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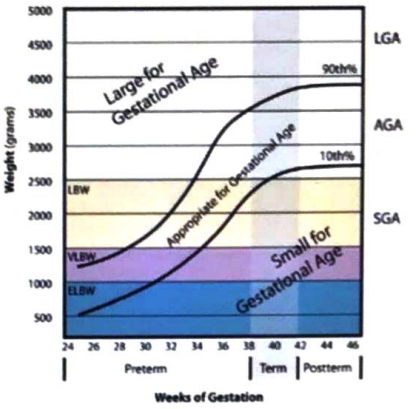
NORMAL NEWBORN

Definitions 00:00:28

Newborn period : First 4 weeks after birth.
(First week is early newborn, next 3 weeks are late newborn period).
Average birth weight in India is 2.9Kgs.
< 2.5 Kgs : Low birth weight.
< 1.5 Kgs : Very low birth weight.
< 1 Kg : Extremely low birth weight.
Big baby/Macrosomia :
> 4 Kgs at time of birth.



Birth weight should always be interpreted with gestational age using intrauterine growth chart (percentile chart).
Lower reference curve is 10th percentile.
Upper reference curve is 90th percentile.

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Any value above 90th percentile is Large for Gestational Age (LGA).
Baby weight between 10th percentile and 90th percentile is Appropriate for Gestational Age (AGA).
Baby weight < 10th Percentile is Small for Gestational Age (SGA).

Small for gestational age 00:04:25

Types :

- Normal variant/Constitutional SGA (more common).
- Abnormal variant : IUGR (Intrauterine growth restriction /retardation).
  Always associated with pathology.

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Points to differentiate IUGR from normal variant SGA :

IUGR babies have loose skin folds (at least 3).

Placenta/umbilical cord appears thin.

Head circumference >> chest circumference (difference > 3 cm). Normal difference between head and chest circumference is 3 cm.

Types of IUGR :

	Maternal cause (more common)	Fetal cause
	Disorders of utero placental insufficiency like Gestational HTN	Genetic defects, anomalies.
Onset	Late (2nd or 3rd trimester)	Early (1st trimester)
Effect	Brain sparing (because majority of bood supply is directed towards the brain). Head size is normal & body is small, called asymmetric IUGR.	No brain sparing (because of early onset). Every part of the body undergoes growth restriction : Symmetrical IUGR.

$$\text{Ponderal Index (PI)} = \frac{\text{weight (g)}}{\text{length (cm)}^3} \times 100$$

PI > 2 : Symmetric IUGR.

PI < 2 : Asymmetric IUGR.

Large for gestational age (LGA)

00:12:21

Birth weight > 90th percentile.

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Causes :

1. Constitutional LGA/ normal variant .
2. Infant of Diabetic mother (gestational diabetes).

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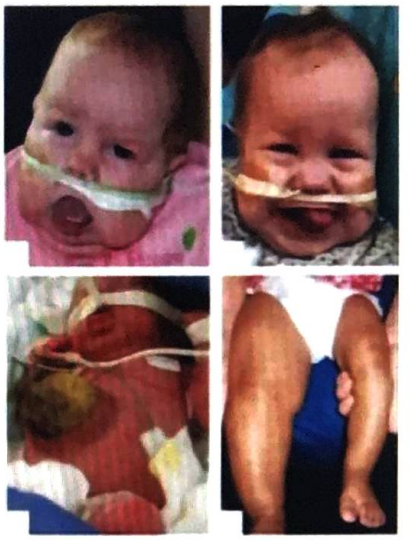
3. SOTOS syndrome (cerebral gigantism) :
   Child has Intellectual disability,
   Premature tooth eruption,
   Developmental delay.

4. Beckwith-Wiedemann syndrome.
   Macroglossia.
   Anterior abdominal defects like Omphalocele & Umbilical hernia.
   Hemihypertrophy.
   Increase risk for Wilms's tumor.



Baby with Beckwith-Wiedemann syndrome.

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Assessment of maturity 00:16:25

Newborn are divided into Pre-term, Term and Post-term babies according to the gestational age.
GA < 36 weeks + 6 days : Pre term baby.
GA 37 weeks to 41 weeks + 6 days : Term baby.
GA > 42 weeks : Post term baby.
Gestational age is calculated from LMP.

Post natal assessment of Gestational age is done using Expanded New Ballard Score (ENBS).
Criteria :

1. Physical maturity.
2. Neuromuscular maturity.

Range of gestation : 20 weeks to 44 weeks.
Total Score range : -10 to +50.
Accurate to ± 1 week.

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Neuromuscular maturity :

Score	-1	0	1	2	3	4	5
Posture							
Square window (wrist)	> 90°	90°	60°	45°	30°	0°	
Arm recoil		180°	140–180°	110–140°	90–110°	<90°	
Popliteal angle	180°	160°	140°	120°	100°	90°	< 90°
Scarf sign							
Heel to ear							

Physical maturity

Skin	Sticky, friable, transparent	Gelatinous, red, translucent	Smooth, pink, visible veins	Superficial peeling and/ or rash ; few veins	Cracking, pale areas; rare veins	Parchment, deep cracking; no vessels	Leathery, cracked, wrinkled
Lanugo	None	Sparse	Abundant	Thinning	Bald areas	Mostly bald	
Plantar surface	Heel–toe 40–50 mm : –1 < 40 mm : –2	> 50 mm, no crease	Faint red marks	Anterior transverse crease only	Creases anterior 2/3	Creases over entire sole	
Breast	Imperceptible	Barely perceptible	Flat areola,no bud	Stippled areola, 1–2 mm bud	Raised areola, 3–4 mm bud	Full areola, 5–10 mm bud	
Eye/ear	Lids fused loosely : – 1 tightly : – 2	Lids open ; pinna flat ; stays folded	Slightly curved pinna; soft ; slow recoil	Well curved pinna ; soft but ready recoil	Formed and firm, instant recoil	Thick cartilage, ear stiff	
Genitals (male)	Scrotum flat, smooth	Scrotum empty, faint rugae	Testes in upper canal, rare rugae	Testes descending, few rugae	Testes down, good rugae	Testes pendulous, deep rugae	
Genitals (female)	Clitoris prominent, labia flat	Clitoris prominent, small labia minora	Clitoris prominent, enlarging minora	Majora and minora equally prominent	Majora large, minora small	Majora cover clitoris and minora	

Maturity Rating

Score	Weeks
-10	20
-5	22
0	24
5	26
10	28
15	30
20	32
25	34
30	36
35	38
40	40
45	42
50	44

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Differentiating points based on appearance :

	Preterm	Term
Posture	Extension. Less tone.	Flexion. Good tone. (attitude of universal flexion)
Scarf sign	Positive (Flexed elbow crosses midline) : Less tone.	Negative (Flexed elbow does not cross midline) : Good tone.
Breast buds/ nodule	<5 mm	>5 mm
External genitalia : male	Scrotum : Appears light very less/absent rugae. Testis not palpable (as it descends by 36 weeks).	Scrotum : Pigmented. Rugae ++. Testis palpable.
External genitalia : Female	Labia majora and minora are visible.	Labia majora is visible (it covers labia minora).
Ear recoil	Slow (elastic cartilage not well developed).	Fast (well developed elastic cartilage).
Lanugo hair (fetal hair)	Abundant.	Sparse/absent.

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Points to identify post term baby :

1. Meconium staining over nails, fingers & body.
   After 42 weeks, anal sphincter opens.
2. Overgrown fingernails.
3. Wrinkling of skin (decrease in amniotic fluid).

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ROUTINE NEWBORN CARE

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Routine newborn care

00:29:14

1. At birth : 5 cleans.

- Clean hands & wear gloves.
- Clean surface (towel used to receive babies should be clean and warm).
- Clean blade/scissors (for cutting umbilical cord).
- Clean cord clamp.
- Clean cord (It should be kept dry).

Delayed cord clamping :

- Delayed clamping by 30-60 seconds.
- Results in increased blood flow via placenta which decreases risk of anemia in later life.
- Preterm babies : Decreases risk of intraventricular hemorrhage.

2. Prevention of hypothermia :
   - Delivery room temperature at 25°C.
   - No free draft of air in the delivery room.
   - Facilitate skin to skin contact of baby and mother.

3. Breastfeeding : Should be started as early as possible after birth.

4. Rooming-in : To keep the baby and the mother together, facilitates bonding between mother and baby. Also helps in breastfeeding.

5. Prophylaxis at the time of birth :
   - Vitamin K : Activates clotting factors 2,7,9,10. Given IM in anterolateral aspect of thigh.

Active space

- Dose : 1 mg. If weight < 1 kg (ELBW) : 0.5 mg.
- Prevents hemorrhagic disease of newborn.

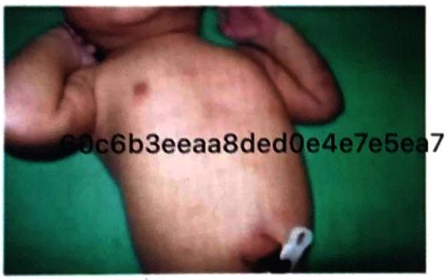
Normal observation in newborn 00:35:04

Vital signs :

- Temperature : 36.5°C to 37.5°C.
- Heart rate : 110-160/min.
- Respiratory rate : 40-60/min.
- Blood pressure : 60/40 mm hg.
- CFT (capillary filling time) : measure of circulation in the body. Checked over bony prominence. Preferred site is over sternum. Normal : < 3 secs.

Normal findings in newborn :

1. Skin and mucosa :



Erythema toxicum

Erythema toxicum is a misnomer. Now called erythema neonatorum.

Normal skin findings	Erythema toxicum/ neonatorum (most common)	Pustular melanosis
Appearance	Red and yellow papulo-pustular lesions. Appears after 24hours.	Hyperpigmented patches + pustules. Appears within 24hours.
microscopy	Eosinophils ++	Neutrophils++
Treatment	Reassurance	Reassurance

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2. Effect of maternal estrogen :

- White discharge per vagina (mucus from cervix)
- Breast engorgement.
- Bleeding per vagina (due to withdrawal of maternal estrogen leading to endometrial sloughing).

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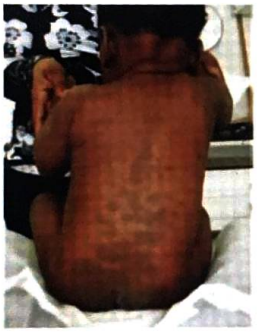
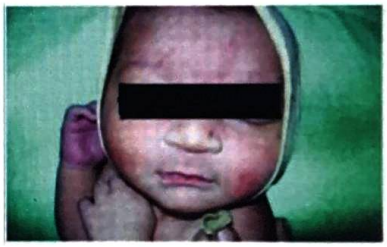
3. Epithelial inclusion cyst/Retention cyst (Appears white)

- Milia : Around the nose and face. (milk spots).
- Gums : Bohn's nodules (white spots).
- Palate : Epstein pearls.

Milia (milk spots)



4. Mongolian spots
Hyperpigmented macules. Very common in Indian and Asian people.



5. Salmon patch/strawberry angioma/stork bite/nevus flavus
Bright red coloured lesion. Also seen in the forehead and nape of neck.

Other normal findings :

- Small Subconjunctival/retinal hemorrhages (while passing through birth canal).
- Hymenal skin tags in female babies.
- Physiological phimosis in male babies.

Reassurance is the only treatment required for all normal new born findings.

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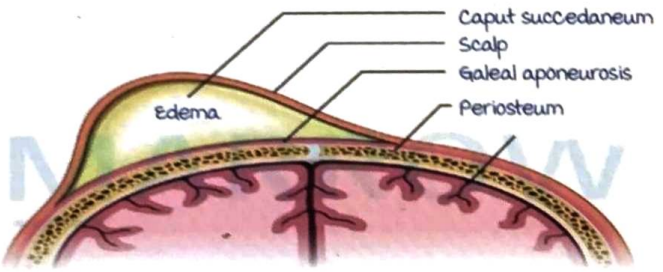
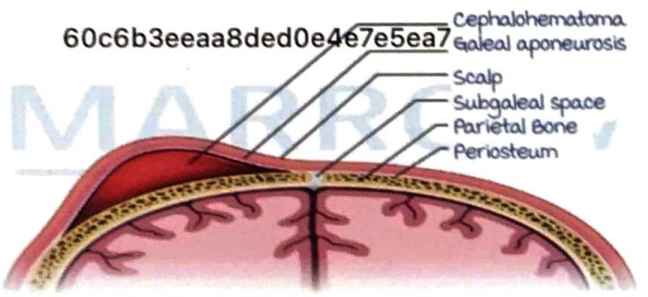
Head swellings in newborn

00:47:35

	Caput succedaneum (more common)	Cephalohematoma
Location	Subcutaneous plane. Superficial swelling in scalp. Diffuse.	Sub periosteum (common : Parietal bone) Deep swelling, localised (limited by sutures).
Reason	Prolonged labour → Congestion of scalp veins → Edema	Instrumental delivery → Trauma to skull
Content	Fluid	Blood
Appear-ance	At birth/soon after birth.	Slowly increases over a period of 12-24hours after birth.
Disap-pearance	48-72 hours.	3-6 weeks.
Associa-tions	Negative	5-25% linear skull fractures, jaundice (from haem break down forming bilirubin)



Caput succedaneum



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Cephalohematoma

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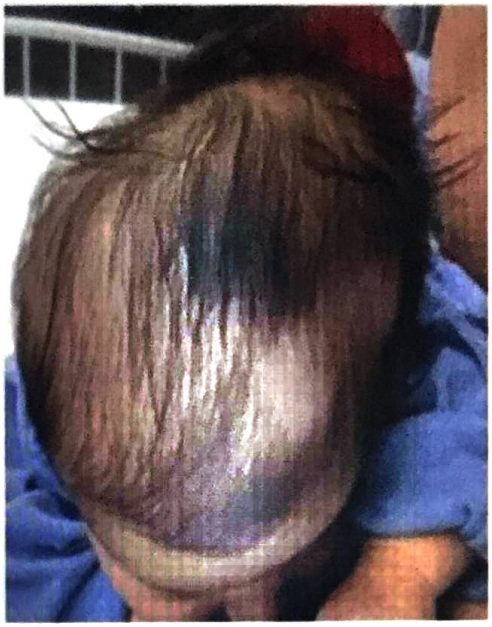
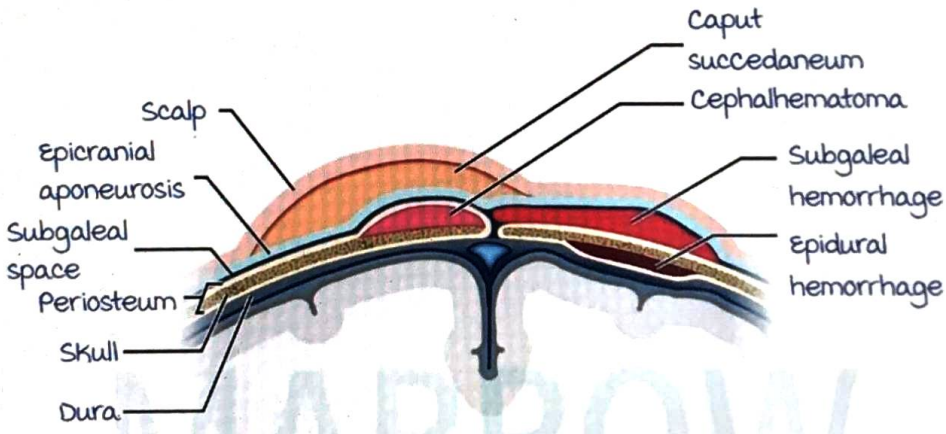


Subgaleal hemorrhage (most severe) :

- Beneath galea aponeurosis.
- Associated with vacuum assisted delivery.
- Diffuse accumulation of blood.
- Hypovolemia →shock.
- Pallor (due to decreased circulating volume).
- Jaundice (lysis of RBCs).



MARROW

Head swellings summary :

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Superficial, diffuse collection of fluid	Caput succedaneum
Superficial, diffuse collection of blood	Subgaleal hemorrhage
Deep, localized collection of blood	Cephalohematoma

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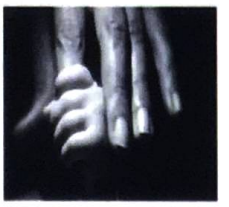
Neonatal reflexes

00:59:27

Also called primitive/immature reflexes.

Asymmetric tonic neck.

Rooting reflex.



Palmar grasp reflex.

Forward Parachute Reflex (Protective extension reaction forward)

Primitive reflexes : Disappear after brain matures.
Generally present during the neonatal period but with few exceptions.

Reflex	Onset	Fully developed	Duration
Palmar grasp	28 weeks of gestation.	32 weeks of gestation.	2-3months after birth.
Rooting reflex (attachment & sucking while breast feeding)	28-32 weeks of gestation.	34-36 weeks of gestation.	Less prominent 1 month after birth.
moro reflex (seen only in term babies)	28-32 weeks of gestation.	37 weeks of gestation (term).	5-6 months after birth.
Asymmetric tonic neck	35th week of gestation	1 month after birth	6-7 months after birth
Parachute	7-8 months (post natal)	10-11 months	Persists throughout life

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Active space

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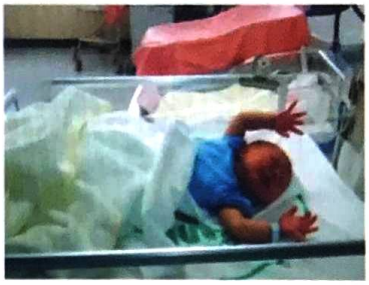
Components of moro reflex :
Extension & abduction followed by flexion & adduction ± cry.



moro reflex

Abnormalities :

Absent moro reflex :
Anomalies like anencephaly.
Hypoxic ischemic encephalopathy (HIE stage 3).

Unilateral moro reflex :
Nerve injury (brachial plexus injury e.g. Erb's palsy).
Bony injury like fracture's /dislocation (m/c : Shoulder).
most common fracture at time of birth : Fracture clavicle.

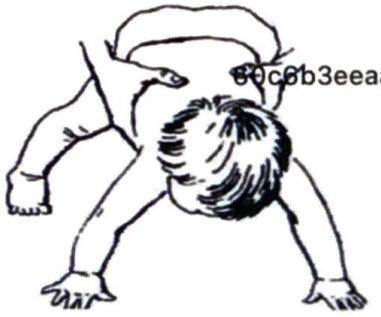
Persistent moro even after 6 months : Immaturity of brain (cerebral palsy).

Asymmetric tonic neck reflex :
If head of the baby is turned to one side, the upper limb on the same side willl extend, while the other upper limb becomes flexed.

Rooting reflex : Stimulation of the baby's mouth causes the mouth to turn in the direction of the stimulus.

Reflexes appearing after birth/ Infantile reflex 01:11:13

Mnemonic : SPL (special)
Parachute reflex/.
Protective extension reaction forward.
Prevents head from falling down.



Forward Parachute Reflex

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Symmetric Tonic Neck Reflex (STNR) :

Also called crawling reflex.

Neck extension → Upper limb extension & lower limb flexion.

Neck flexion → Upper limb flexion & lower limb extension.

Appears by 4 - 6 months after birth.

Duration : 8-12 months after birth (helps in crawling).

Symmetrical Tonic Neck Reflex

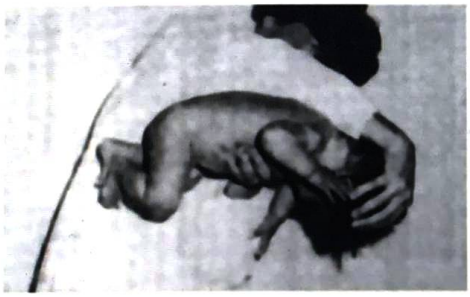
Landau reflex :

On horizontal suspension, flexion of neck → flexion of limbs.

Extension of neck → Extension of limbs.

Appears : 3 months after birth.

Duration : 9 months after birth



Landau reflex

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MANAGEMENT OF LBW BABIES

Care of LBW babies 00:00:30

Reasons for LBW :

1. Preterm babies (developed countries).
2. IUGR/growth restriction (developing countries) → SGA babies (Small for Gestational Age).

m/C problem faced by these babies : Hypothermia.

Temperature regulation in a new born :

Normal : 36.5–37.5 °C.

Site of measurement : Axilla.

(For precise recording : Bulb of thermometer should be placed over roof of dry axilla for a minimum of 3 minutes, while holding the axilla close to the baby's body).

modes of heat loss in a new born:

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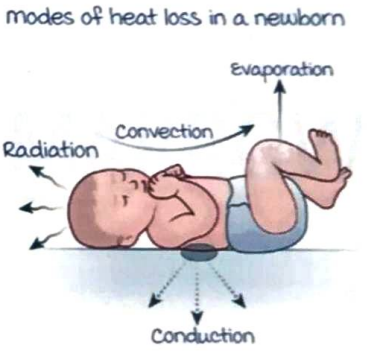
1. Conduction heat loss (baby will lose heat when placed on a cold surface).
2. Evaporative heat loss (sweating).
3. Convection heat loss (air current or air flow around the baby, causing heat loss).
4. Radiation heat loss (heat from baby radiating to the roof/walls of the room).



Most important mechanism of heat loss : Radiation heat loss.

Most important site of heat loss : Head, because it has the largest Body Surface Area (BSA) when compared to the other body parts.

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Hence, baby's head must be covered to prevent excessive heat loss.

LBW babies more prone to hypothermia than normal weight babies :

LBW babies (Preterm)	Normal weight babies (Term)
Greater surface area of head.	Lesser surface area of head.
Extension posture → more heat loss	Flexion posture (protects from heat loss).
Low brown fat.	more brown fat.
Less body fat → Less insulation → Heat loss.	more body fat.
Skin more permeable → Heat loss.	Skin less permeable.

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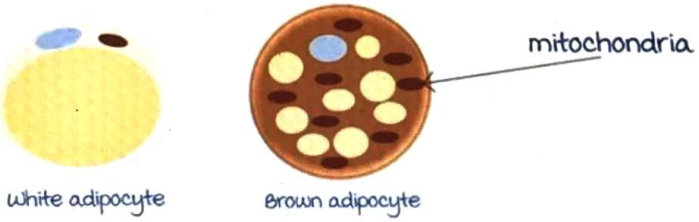
Sources of heat production

00:06:34

Increased physical activity (crying/movements) → Increased heat production.
Cold exposure → Stress → Sympathetic surge (release of sympathetic hormones) → Increased heat production through brown fat.

Brown vs white adipose tissue :



White/ usual adipocyte	Brown adipocyte
One large fat vacuole.	Small fat vacuoles.
Less mitochondria.	more mitochondria (brown structures).
Less vascularity.	more vascularity.
Less nerve innervation.	more nerve innervation (sympathetic nerves).

Brown fat cells : Specialized structures present in new born which helps in heat production.

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Location of brown fat :

1. Nape/back side of neck (MC).
2. Interscapular region.
3. Axillar or groin region.
4. Peri-renal areas.

Non shivering thermogenesis

00:10:35

- mechanism of generation of heat in a new born, without shivering, with the help of brown fat tissue.
- In a fat cell, oxidation of fatty acids takes place, followed by phosphorylation. That is, oxidation couples with phosphorylation (coupling reaction) to produce ATP.

Exposure to cold
↓
Stress
↓
Sympathetic surge
↓
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Release of sympathetic hormones (Norepinephrine : MC)
↓
Uncoupling reaction (oxidation inhibited from phosphorylation)
Diverts oxidation towards heat generation.

Hypothermia (< 36.5°C) :
Normal temperature : 36.5-37.5°C.
Classification :

1. Cold stress (36-36.4°C) : Extremities become cold, trunk remains warm (initial stage).
2. Moderate hypothermia (32-36°C) : Entire body becomes cold.
3. Severe hypothermia (< 32°C) : Entire body becomes cold.

Prevention of hypothermia :
Stable hemodynamically :

- Gently cover the baby (especially the head).
- If LBW/preterm, Kangaroo mother care (skin-skin contact

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with mother → Heat transfer from mother to baby → Protects baby from hypothermia).

Kangaroo Mother Care (KMC)

00:15:40
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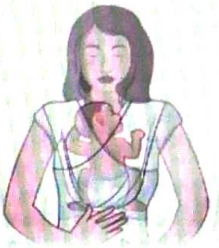


Kangaroo mother care

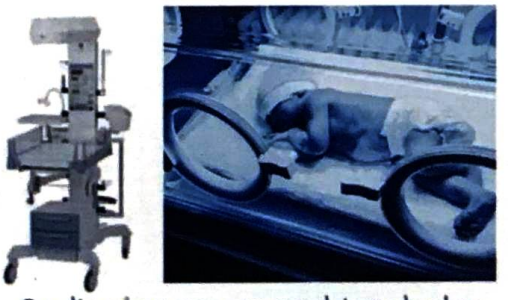
1. Position :
   Posture of baby : Vertical or upright.
   Position of baby's face : Turned to one side.
   Position of legs : Flexed at hip and knees (frog leg posture).
   Head of the baby covered additionally.
2. Feeding : Exclusive breast feeding.
3. Early discharge of baby (due to better care of baby be cause of contact with mother).

KMC duration : Variable (can be upto 24 hrs).
Long duration : most preferred.
Short duration (< 1 hour per session) : Avoided.
KMC can be stopped :

- When baby is of term gestation from birth (if premature).
- When baby weighs > 2.5 kg (if LBW).

Unstable hemodynamically : Managed in NICU (Neonatal ICU).

1. Radiant Warmer (radiation type of heat exchange).
   Heat generated over the surface → Radiates down to the baby.
2. Incubator (Convection type of heat exchange) : Closed chamber.



Radiant warmer and Incubator

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Nutrition of LBW babies

00:22:20

Depends on gestational age :

< 28 weeks of gestation (extremely premature) :

Total parenteral nutrition (TPN) : IV fluids.

- No enteral feeding as gastrointestinal tract is not developed completely.

Preferred IV fluids :

- < 48 hours : 10% dextrose.
- > 48 hours : Isolyte-P.

Amount of IV fluids to be given :

Day 1 : 60-80 ml/kg/24 hours.

↓ Increase by 10-20 ml/kg everyday.

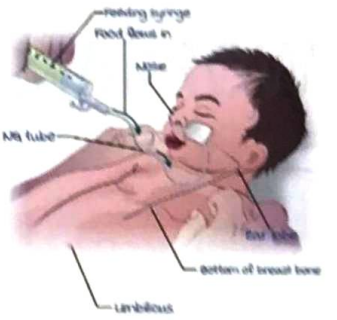
150 ml/kg/24 hours (maximum level) and maintain it.

28-32 weeks of gestation :

- Only suckling burst seen (Suckling + swallowing + breathing coordination not completely developed). Expressed Breast Milk (EBM) given carefully.
- Baby may aspirate EBM if given by mouth directly.
- Therefore EBM given through nasogastric tube/ orogastric tube .
- Length of insertion of NG tube:

Nose to ear lobule to midpoint between xiphoid sternum & umbilicus should be measured and marked then nserted.

Nasogastric tube in newborns



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32-34 weeks of gestation (coordination better developed) :

EBM with a paladai or Katori spoon.

> 34 weeks (completely developed coordination) :

Direct breast feeding.

Active space

Nutritional supplements

00:31:21

Weight	Supplements
All babies	Vitamin D (400 IU/day : Orally for 1 year).
1.5–2.5 Kg	Vitamin D + Iron (2mg/kg/day) : From 6–8 weeks after birth till 1 year. (LBW babies have inadequate iron stores).
< 1.5 Kg (very low birth weight)	Iron + HMF (Human Milk Fortifier). Nutritional sachets continued till baby reaches 40 weeks of gestation after birth. Continue Iron + Vitamin D for 1 year. HMF contains all micro nutrients + minerals + vitamins, except iron. Therefore, iron supplements in addition.

Problems faced by LBW babies (preterm/IUGR babies) :

	Preterm babies	IUGR babies
Neonatal asphyxia	Increased risk.	Increased risk.
Hypothermia	Increased risk.	Increased risk.
Low nutrient reserves	Hypoglycaemia (low glycogen reserves).	Hypoglycaemia (low glycogen reserves).
Kernicterus (brain damage due to bilirubin)	Increased risk. High levels of bilirubin → Easy passage into brain (blood brain barrier is more permeable in preterm babies).	-
Necrotizing enterocolitis (NEC)	Increased risk.	-
Patent Ductus Arteriosus (PDA)	Increased risk.	-
Hematological	Anemia (low Iron stores).	Polycythemia. IUGR → Stress → Intrauterine hypoxia (chronic) → Release of EPO → Increase erythropoiesis → Polycythaemia.

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Active space

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Respiratory	Less surfactant causing Respiratory Distress Syndrome (RDS)	meconium aspiration syndrome (MAS). IUGR → Stress → Release of motilin → Increased peristalsis of intestines → Passage of meconium in-utero → Aspirated by baby during delivery → MAS.

Problems faced by premature babies due to immaturity of organ systems :

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1. Intraventricular haemorrhage (IVH) : < 34weeks.
   - Germinal matrix (group of fragile blood vessels around the ventricles) undergoes involution > 34 weeks.
   - If baby < 34 weeks → Germinal matrix still present → Prone to bleed → Rupture → Bleeding into the ventricles.

2. Increased risk of apnoea of prematurity (absent breathing > 20 seconds or any duration + bradycardia or cyanosis).
   Treatment : Caffeine citrate.

3. Increased risk of developing Sensorineural Hearing Loss (SNHL) : Frequent screening tests done.
   - Oto-Acoustic Emissions (OAE).
   - Automated Auditory Brainstem Response (AABR) : most preferred.

4. Increased risk of developing Retinopathy of Prematurity (ROP), possibly leading to blindness.
   - Either due to immaturity of retina.
   - When exposed to high levels of oxygen therapy (hyperoxia).
   - Prevented by limiting O_a exposure : By continuously monitoring SpO_a (90-95% maintenance).

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NEONATAL RESUSCITATION

Neonatal resuscitation program/NRP 2021 00:00:33

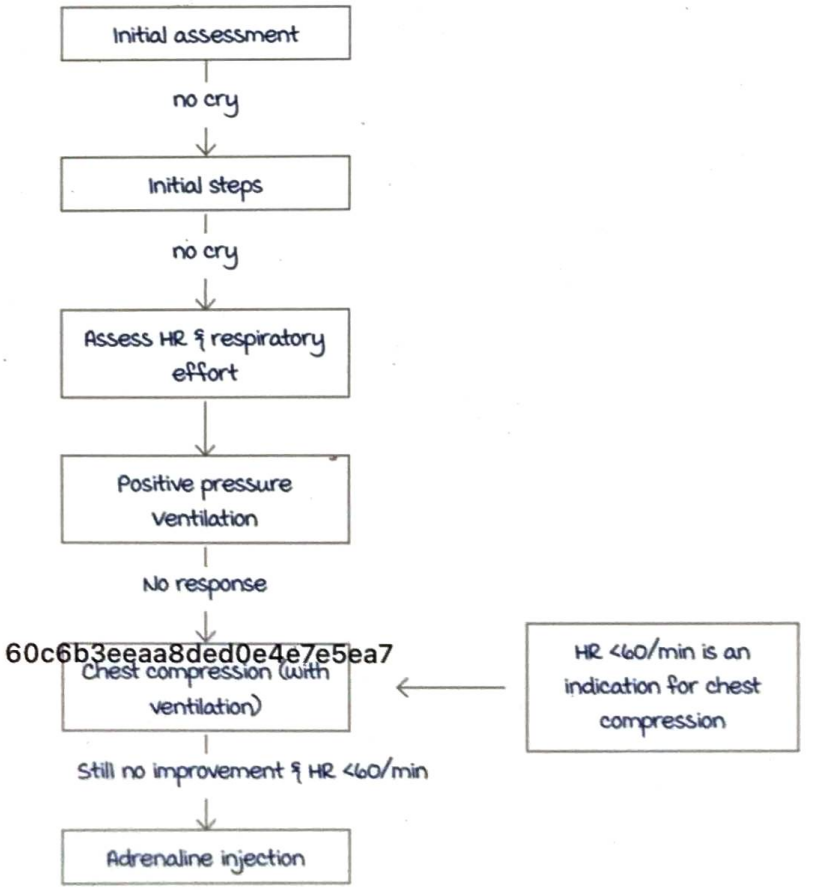
The guidelines for neonatal resuscitation are based on NRP 2021.

In newborn babies, the priority is respiration >> temperature > circulation.
Resuscitation in newborns is not done under room air but under warmer air.

Initial assessment : 4 pre birth questions are asked.

- Gestational age of the baby.
- Amniotic fluid is clear or not.
- Additional risk factors.
- Umbilical cord management plan → Delayed cord clamping by 30 -60 seconds.

Algorithm :



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Initial steps :

- Temperature → Needs to be maintained by putting the baby into the warmer.
- Stimulate the baby by gentle tactile stimulation →
  1. Rubbing the back of the baby.
  2. Flicking the soles of the baby.

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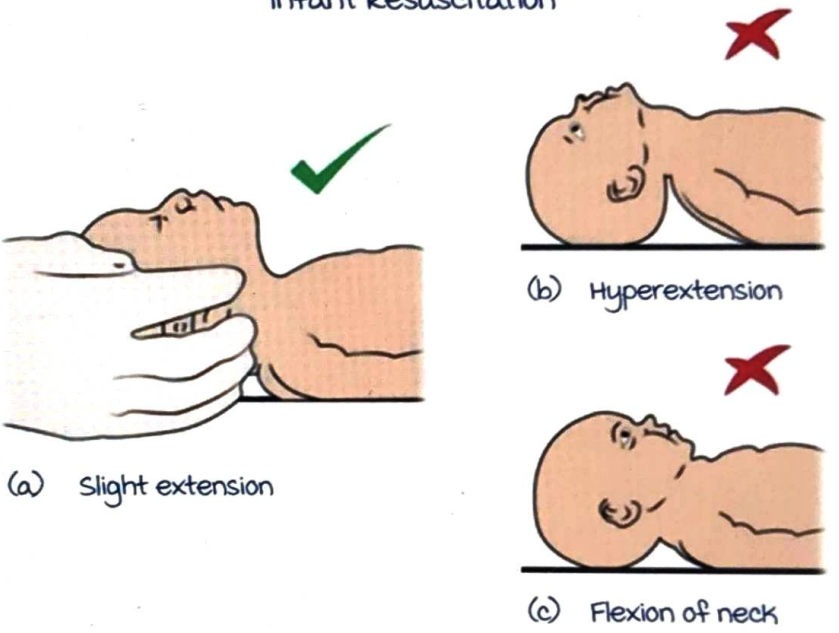
- Position the airway of the baby by slight extension/ sniffing posture.

  Easily done by placing a rolled cotton towel behind the shoulder blades.

Infant Resuscitation



(a) Slight extension

(b) Hyperextension

(c) Flexion of neck

Optional step : Suctioning of the secretions (mouth → then nose) to prevent aspiration (routine suctioning not recommended anymore).

If the baby still doesn't cry after the initial steps, assess for heart rate (to check for hypoxia) and respiratory effort.

Significant hypoxia is a decrease in heart rate : < 100/min (normal : 120–160/min).

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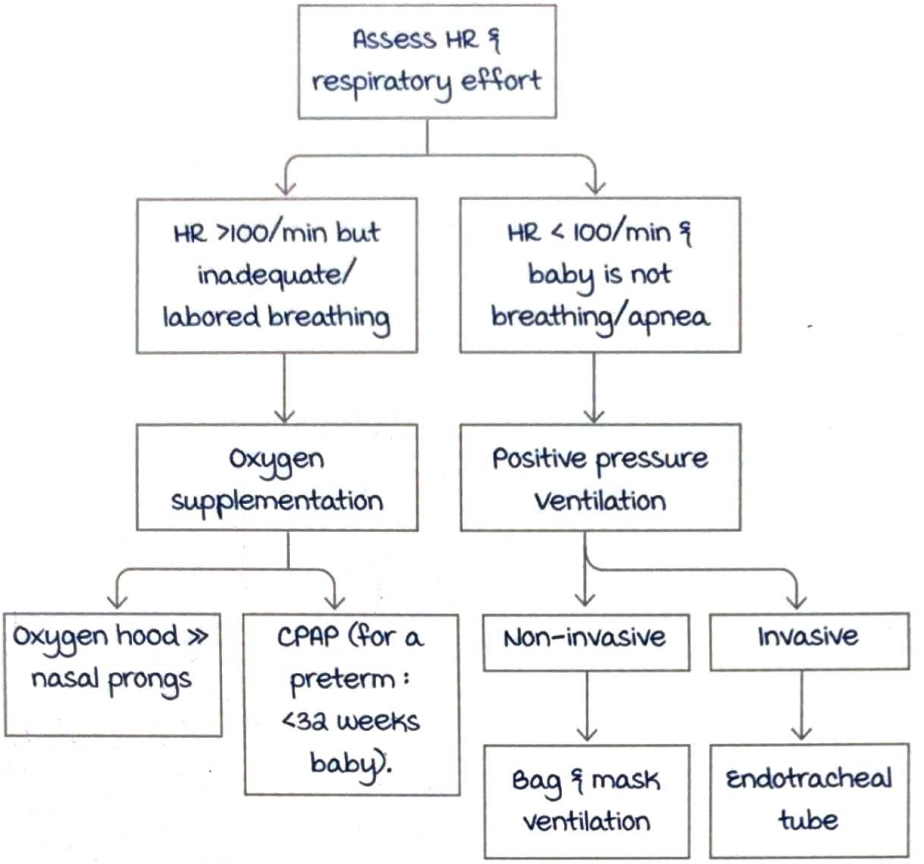

If still no response and heart rate <60/min, chest compressions are indicated.

2 thumb technique :

Site : Lower 1/3rd of sternum.

Avoid xiphisternum.

1/3rd of the antero-posterior diameter should be compressed.

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Chest compressions should be combined with ventilation in a newborn resuscitation.

Active space

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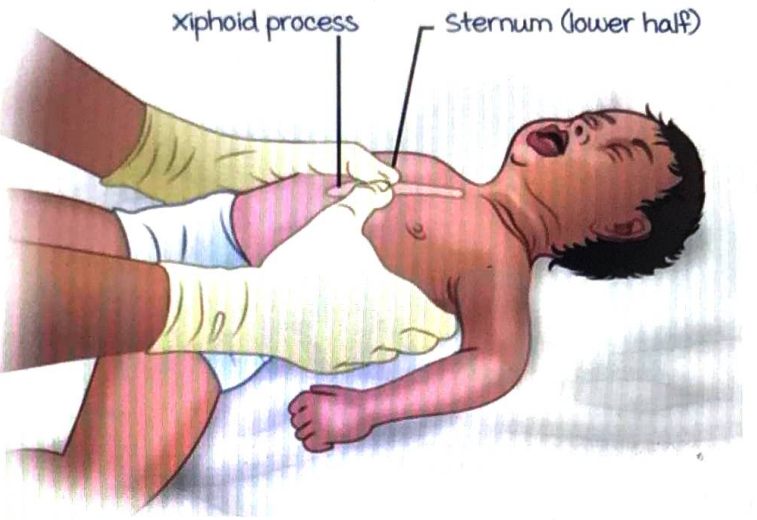
Neonatal Resuscitation – Chest Compression



Ratio of compression : Ventilation → 3:1 → 90 compressions + 30 ventilation in 1 minute.

Ventilation is usually given by an alternate airway like ET ventilation.

Cardiac monitor is necessary at this stage to monitor HR.

No response to chest compressions then,

Adrenaline injection : (1:10000 → strength).

Route → Umbilical vein (IV >> Interosseous route > ETT).

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Dose → IV/Interosseous route → 0.2 ml/kg or 0.02 mg/kg.

Endotracheal route → 1 ml/kg or 0.1 mg/kg.

If even after adrenaline injection, there is no improvement → consider volume expansion : NS 10 ml/kg (not a routine step, usually done as a last effort where definite history of blood loss (e.g. PPH etc.,) present.

Maximum duration of neonatal resuscitation → 20 minutes → call off resuscitation if all steps done & no HR.

Timeline between the initial step to PPV is 1 minute (golden minute of resuscitation).

Timeline between PPV & chest compression initiation → 30 seconds.

Timeline between chest compression initiation & adrenaline injection → 30 seconds.

Active space

Bag & mask ventilation / BMV

00:26:29

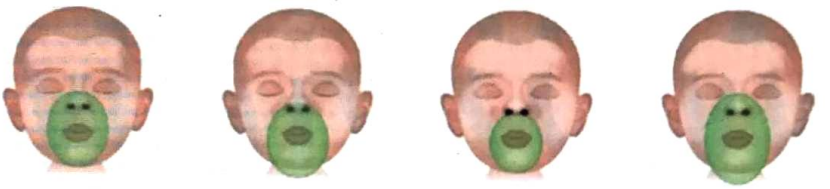
Volume of bag for resuscitation : 240–750 ml.

Response : Increase in HR > 100/min.

If no response → ventilation corrective steps.

- Mask readjustment.
- Reposition head.
- Suction.
- Open the mouth.
- Pressure to be increased.
- Alternate airway (ET).

Correct position of the mask



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Rate of BMV → 40–60/min.

Pressure : First breath → 30–40 cm H_2O.

subsequent breaths → 15–20 cm H_2O (always at lower pressure to prevent trauma to the alveoli).

Recommended FiO_2 → babies born ≥ 35 weeks : Room air (21% O_2).

Babies born < 35 weeks : 21–30% O_2.

Further titration of oxygen depends on SpO_2 response.

Target SpO_2 :

1 min	60–65%
2 min	65–70%
3 min	70–75%
4 min	75–80%
5 min	80–85%
10 min	85–90%

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Excessive oxygen can lead to deleterious effects on newborns → Retinopathy of Prematurity/ROP → if untreated, can lead to blindness.

SpO_a is usually checked by placing the pulse oximeter on the right hand.

Target spo_a at 3 minutes → 70 – 75%.

Absolute contraindication → congenital diaphragmatic hernia (intestines are already compressing the lungs and with BMV → air enter both trachea and oesophagus → pushing more air in intestines leading to more compression of lungs.

Endotracheal intubation 00:36:12

Before ET, laryngoscopy with straight blade (0 for preterm & 1 for term babies) needs to be done to visualise glottic region.

Uncuffed tube will be preferred in new-borns (pressure induced tracheal necrosis can occur if cuffed ET is used).

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Inner diameter of ET → < 1 kg child : 2.5 mm diameter.

1–2 kg → 3 mm.

>2kg → 3.5 mm.

Length (cm) of insertion of ET depends on the age of the child.

Length = 6 + weight of child (kg).

Best way to confirm position of ET → End tidal CO_a with capnography.

Active space

INFECTIONS IN NEONATES

Types of infections 00:00:08

Superficial infections & Systemic infections.

Superficial infections :

1. Omphalitis (infection of the umbilical cord) :

Can present as redness/induration/pus discharge around umbilicus.

If untreated, causes systemic infection (sepsis).

Management depends on :

Size of induration	Presence of sepsis	Treatment
Less than 1 cm	No	Topical 0.5% gentian violet 4 times a day
more than 1 cm	Yes/no	Systemic antibiotics (treat like sepsis)

2. Oral thrush :

By candida species.

Differentiated from other white oral lesions by :

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- Difficulty in wiping the lesion.
- Presence of hemorrhagic spots after forceful removal.

Treated with topical Nystatin or Clotrimazole 4 times a day till all the lesions are resolved.

3. Conjunctivitis (ophthalmia neonatarum) :

Acquired during delivery of the baby along the infected birth canal.

	Gonococci	Chlamydia
Organism	Neisseria gonorrhoea	Chlamydia trachomatis
Time of presentation	Day 3–7	From 5th day of delivery
Discharge	Copious and purulent	Scanty
Treatment	IV Ceftriaxone/ Cefotaxime for 7–10 days	Oral Azithromycin for 3 days/ Erythromycin for 14 days

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Gonococcal conjunctivitis is treated as an emergency. Blindness and systemic infection can occur if untreated. 0.5% Erythromycin eye ointment is used as prophylaxis against ophthalmia neonatarum.

Systemic infections

00:08:26

Neonatal sepsis (in the 1st month of life):
2nd most common cause of neonatal mortality after prematurity.
Diagnosed by presence of clinical features of sepsis along with bacteremia (presence of bacteria in blood as detected by blood culture).

Causative organisms	
India	Klebsiella > Staphylococcus aureus > E.coli
Worldwide	Group B Streptococci

	Early onset sepsis	Late onset sepsis
Onset	Within 72 hours of life	After 72 hours of life
Source	From the mother (in the uterus/passage via birth canal)	Hospital acquired infection (Nosocomial)
Risk factors	• Chorioamnionitis (high fever in mother + foul smelling liquor) • Duration of ROM >24hours (>72 hours: very high risk). • PPROM (any duration). • ≥ 3 unclean vaginal examinations.	• Unclean hands of health personnel (most important preventable cause). • Preterm/low birth weight babies. • Absence of breast feeding.
Type of infection	Pneumonia	Septicaemia. Meningitis.

Active space

Diagnosis of neonatal sepsis 00:15:55

Any sick baby, suspect sepsis & start iv antibiotics.
No single most important clinical feature is diagnostic.
Early features : Altered feeding pattern, lethargic baby.
Reliable feature : Hypothermia (more reliable in preterm babies than fever).
Manifestations of affected organ system like respiratory distress, seizures etc.
Management :

1. Gold standard : Blood culture (takes 48-72 hours to get report)
   Start empirical antibiotics.
2. Screen for sepsis using 5 components (Presence of any 2 is significant)

Components	Abnormal value
Total leukocyte count	< 5000/cu mm (immature immune system)
Absolute neutrophil count	< 1800/cu mm
Immature/total neutrophil ratio	≥ 20% immature neutrophils or Ratio ≥ 0.2
Micro ESR	≥ 15 mm/hr
CRP (C-Reactive Protein)	≥ 1 mg/dl

Sepsis screening has negative predictive value (helps rule out infection).
If screening is negative : Absence of disease.
If screening is positive : Do a blood culture to confirm sepsis.

Role of C-reactive protein 00:22:56

CRP raises only 6-12 hours after infection. Peaks at 24 hours.
Disadvantage : Negative in early infection.
Newer and better marker of inflammation : Procalcitonin.
Raises 4-6 hours after infection and peaks at 24 hours.
Procalcitonin is not routinely used in sepsis screening.

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Other investigations :

- Chest x ray (in case of suspected pneumonia).
- Lumbar puncture : In all symptomatic babies with suspected/confirmed sepsis.

(Asymptomatic babies : Presence of risk factors – Lumbar puncture is not required).

Management :

Empirical antibiotics

Gram positive		Gram negative
Penicillin group : Ampicillin	+	Aminoglycoside : Amikacin/ Gentamicin

No response to treatment by 48 hours/severe infection such as meningitis, septic shock.

↓

Add 3rd generation cephalosporin (Ceftriaxone/Cefotaxime)

Suspected Staphylococcus aureus infection : Add Cloxacillin.
In case of MRSA : Add Vancomycin.

In NICU with high incidence of resistant strains :
Cefotaxime/PIPTAZ/Ciprofloxacin + Amikacin.

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Duration of antibiotic therapy in neonatal sepsis :

Diagnosis	Duration
Culture negative sepsis (screening positive and/or clinical course consistent with sepsis)	5–7 days
Blood culture positive without meningitis	14 days
Meningitis	3 weeks
Ventriculitis (Diagnosed using MRI)	4–6 weeks

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BIRTH ASPHYXIA & HYPOXIC ISCHEMIC ENCEPHALOPATHY & NEONATAL SEIZURES

Perinatal asphyxia

00:00:14

Impaired gas exchange in the fetus during pregnancy or due to complications of labor.

Leads to :

- Hypoxia.
- Hypercarbia.
- Lactic acidosis : main feature to diagnose asphyxia.
  Umbilical arterial blood pH <7.

WHO definition of birth asphyxia :
It is a condition that should be considered in any newborn baby wherein the baby fails to initiate breathing or not able to sustain breathing at the time of birth.

Consequences of birth asphyxia : Due to hypoxia.

Organ	Clinical condition
Brain	Hypoxic Ischemic Encephalopathy (HIE).
multi organ failure : Kidney (m/c) after brain.	Acute Tubular Necrosis (ATN) in proximal tubule (most sensitive).
Anaerobic metabolism in tissues	Lactic acidosis.

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Low APGAR score is seen in babies with birth asphyxia : < 7/10.

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Hypoxic ischemic encephalopathy

↓

Hypoxia : Decreased oxygenation to tissues

↓

Ischemia : Hypoperfusion : Inadequate bloody supply to tissues.

↓

Encephalopathy : Brain injury

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HIE :

Leading cause of neonatal brain injury worldwide.

Associated with increased risk of neonatal mortality.

Surviving babies may have neurodevelopmental sequelae like Cerebral Palsy (CP), intractable seizures, & low IQ.

Sarnat & Sarnat staging

00:05:54

Features	Stage 1	Stage 2	Stage 3
Consciousness	Normal/ hyperalert	Lethargic	Comatose
Reflexes (moro, Sucking)	Easily elicitable (hyper active)	Sluggish	Absent
Seizures	-	+++ in first 24 hours	- (no brain activity)

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Autonomic involvement	Sympathetic overactivity	Para sympathetic overactivity	
Pupil size	mydriasis	miosis	Unequal : Poorly/not reactive to light.
Heart Rate	Increased	Decreased	Variable
Posture	Normal	Flexion	Decerebrate
EEG	Normal	Low voltage complexes. Seizure spikes seen.	Burst suppression. Isoelectric pattern.
Duration	< 24 hours	24 hours – 14 days	Several days to weeks
Outcome	99% : Normal	80% : Good outcome. 20% : Sequelae.	50% : Die. 50% : Sequelae.

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Normal posture : universal flexion resisting passive extension.
Decerebrate posture : All limbs in extension.
In stage 2, 20% of babies develop neurological sequale, while in stage 3, all surviving 50% develop cerebral palsy as sequlae.

Child will not always progress from stage I and can present in any stage.
Sarnat & Sarnat staging is also indicative of the prognosis.
HIE is the leading cause of neonatal seizures.

Neurological sequelae depend on the area of brain affected by HIE.
Factors that determine area affected :

- Blood supply of brain.
- Gestational age.
- metabolic demands of different parts of the baby's brain.

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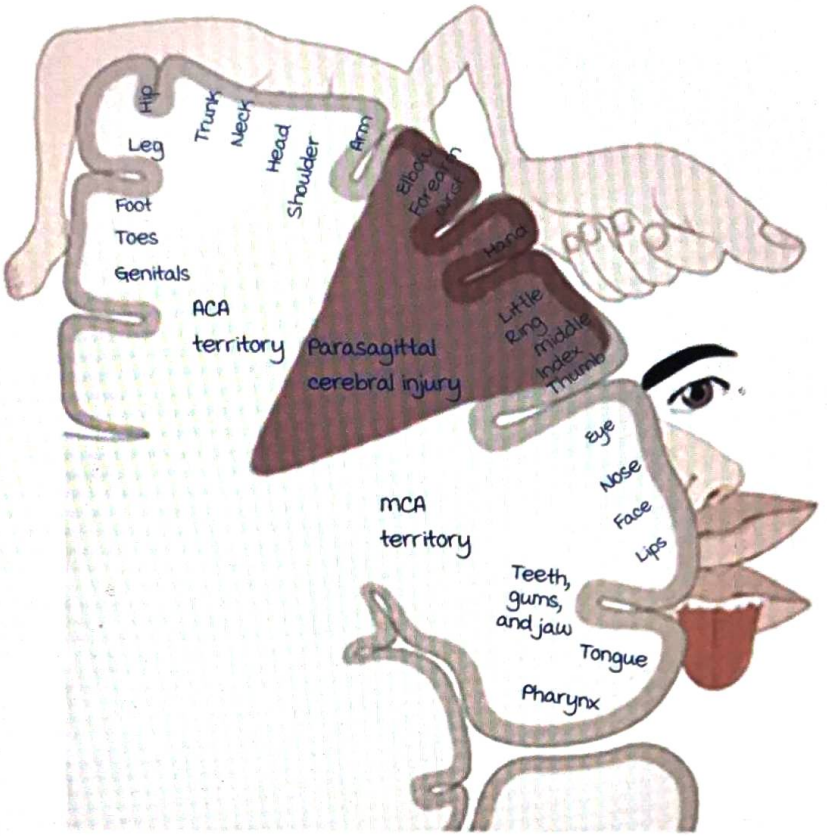
Parasagittal infarct

00:13:35



Parasagittal cerebral injury

Brain injury in term babies due to asphyxia.
Water shed area supplied between anterior & middle cerebral artery.
Motor cortex & subcortical areas are affected.
Initially, upper limbs are predominantly affected.
All 4 limbs will be eventually affected : Spastic quadriplegia cerebral palsy (CP).

Periventricular leukomalacia :
Brain injury in preterm babies.
Affects motor distribution of both lower limbs : Spastic diplegia CP.

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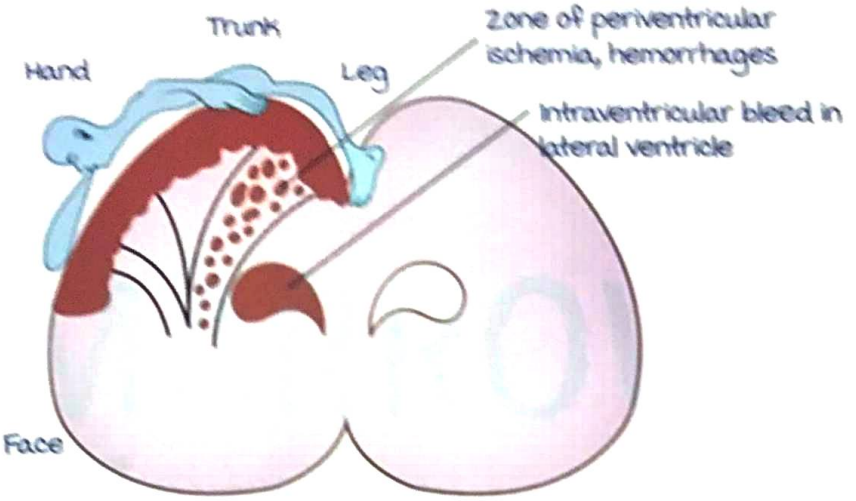

Zone of periventricular hemorrhages

Other patterns of ischemic brain injury in term babies :

Focal ischemic necrosis :
Refers to only one part of brain being affected.
Associated with focal seizures & hemiparesis (upper & lower limbs on one side being afffected).

Selective neuronal necrosis :
Particular areas of brain are commonly affected.
Deep nuclei of brain : Putamen of basal ganglia (extrapyramidal symptoms), Hippocampus.
Purkinje cells of cerebellum : Ataxia, vertiga.
Many areas affected together : CP with low IQ.

Neuroimaging 00:19:25

Screening :
Neurosonogram : Ultrasonogram through fontanelles.
Only small part of brain is visualized.
Main role : To rule out significant intracranial hemorrhages.

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Definitive : MRI of brain.
In early stages : Diffusion Weighted Imaging (DWI) shows abnormality (early changes) within few hours after birth.

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Treatment of HIE :
Cannot completely cure, only supportive management.
Maintain blood sugar, temperature and administer adequate fluids.
aEEG : Amplitude integrated EEG for bedside monitoring of seizure activity in a newborn.
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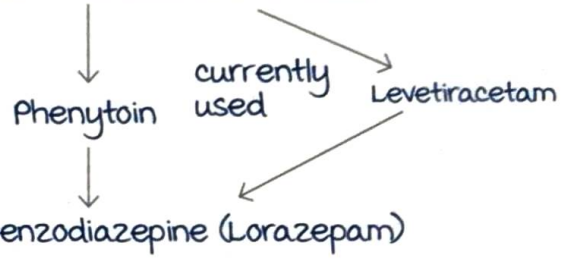
Only 1 – 2 electrodes used (conventional EEG has 16 electrodes)
Conventional EEG needs expert evaluation.
Facilitates continuous monitoring of seizure activity.
Helps in detection of subclinical seizure activity in ICU.
Screening test only. Diagnostic test : Conventional EEG.

Treatment of seizures : Phenobarbitone (DOC)



Therapeutic hypothermia 00:23:43

Inducing hypothermia by placing ice packs or cooling crystals around the baby's body : Whole body cooling.
Only around the head : Selective head cooling.
Preferred : Whole body cooling > selective body calling.
Whole body cooling offers uniform cooling & better outcomes.
Temperature maintained : 33.5 °C (33 – 34°C).

Mechanism of action :
Decreases neuronal injury by decreasing neuronal apoptosis.
Decreases production of free radicals & nitric oxide.
Decreases production of excitatory amino acid glutamate.
Decreases incidence of seizures.

Criteria for therapeutic hypothermia :
Post menstrual age : > 36 weeks of gestation.
Birth weight : ≥ 2 kg.
Stage 2/Stage 3 HIE.
Should be started within 6 hours of birth. Duration : 72 hours.

Active space

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Types and Etiology of Neonatal Seizures 00:29:48

Types :

1. Subtle
   - m/c type in children.
   - Minimal manifestations
     - Ocular movements (Example : Deviation of eye or continuous blinking of eyes for few seconds)
     - Orofacial lingual movements (Example : Continouschewing).
     - Limbs (Example : cycling movement).

   Seizure generated impulse is not transmitted rapidly in the immature brain.

2. Clonic
   - Focal clonic : Best prognosis.
3. Tonic
4. Myoclonic
   - Worst prognosis.

Etiology :

1. HIE (m/c) : 50 – 60% within 12 hours of 1st day of life.
2. Metabolic causes :
   - Hypoglycemia.
   - Hypocalcemia.
   - Hypomagnesemia.
3. Infections :
   - Sepsis.
   - TORCH infections.
4. Intracranial hemorrhage :
   - Preterm : Intraventricular hemorrhage.
   - Term : Subarachnoid hemorrhage (Good outcome).
5. Developmental defects :
   - Congenital anomalies (example anencephaly)

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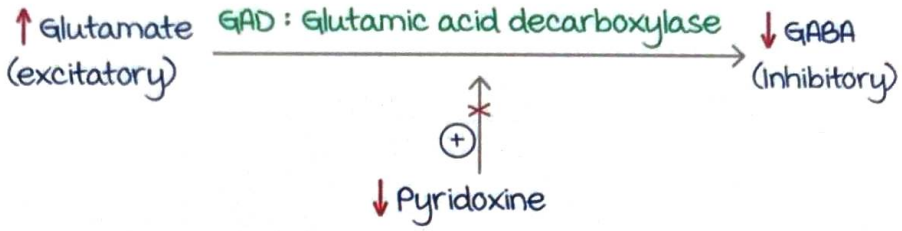
6. Pyridoxine deficiency/ dependent seizures.
   - Rare cause of neonatal seizures.
   - Typically manifests as refractory seizures.



Management of Neonatal Seizures 00:30:53

TABC (Temperature, airway, breathing, circulation)

Correction of hypoglycemia and hypocalcemia

Antiepileptic drug therapy :

First line : Phenobarbitone IV.

Second line : Phenytoin followed by Benzodiazepines.

Refractory seizures : Trial of pyridoxine.

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NECROTIZING ENTEROCOLITIS

Introduction

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00:00:18

Parts of the intestine affected :

- Terminal ileum : most common.
- Proximal colon (ascending colon).

Considered as a gastrointestinal emergency in a neonate. Characterized by inflammation due to bacterial colonization of the intestine, leading to necrosis of that part of the intestine, if left untreated.

Risk factors :

- Prematurity.
- Top feeds (feeding with milk, other than breast milk : cow milk/ formula milk).
  1. Breast milk is protective because it lowers the gastric pH, making it more acidic, which prevents bacteria colonization by pathogenic bacteria like E. coli, Klebsiella.
  2. It also promotes peristalsis of the intestine.
  3. It contains substances like lactoferrin, which protects against the growth of bacteria like E. coli.
  4. Increases normal gut flora.
- Birth Asphyxia.
- Maternal abuse of cocaine.
- Usage of H_a blockers in the neonate.

Pathogenesis :

- Prematurity → Immature GIT
- Top feeds → Bacterial Colonization

] Inflammation leading to necrosis.

- Asphyxia → Vasoconstriction of the splanchnic vessels → predisposed to necrosis → NEC.
- Cocaine abuse → Vasoconstriction of the splanchnic vessels → predisposed to necrosis → NEC.

Active space

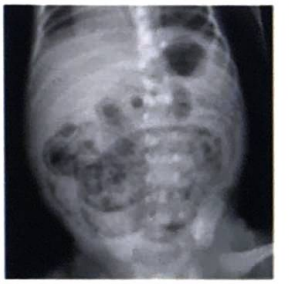
Leave Feedbac

- Usage of H_a blockers → Decreases the HCL amount in stomach → Increases pH of gastric wall → No protection from bacterial colonization.

Features of NEC

00:05:25

Modified Bell's Staging :

Stage	Features	Imaging
Stage I. (initial/ beginning). NEC suspected.	• Non specific abdominal features (distension/ vomiting). • Systemic manifestations : Fever, bradycardia or apnea. Stage IA : Occult blood in the stool. Stage IB : Gross blood in the stool.	Normal X ray
Stage 2. Definite NEC.	Stage 2A : Absent bowel sounds.	X-ray abdomen : Pneumatosis intestinalis (air in the wall of the intestine). 
	Stage 2B : Bowel sounds (-) + Abdominal wall edema. Lab triad : Thrombocytopenia + Hyponatremia + metabolic acidosis.	X-ray abdomen : Pneumatosis portalis (air in the portal vein).

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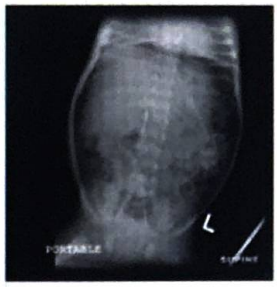
Stage 3 Severe NEC : Complicated/ Advanced NEC.	Stage 3A : Definite evidence of peritonitis (erythema of the abdominal wall + Induration + Tenderness of the abdominal wall) + Definite Ascites. Stage 3B : Intestinal perforation (Air collecting in the peritoneal cavity). Surgical Gastrointestinal Emergency of Neonates.	X-ray abdomen : Pneumoperitoneum (Air or gas under diaphragm). 

Management of NEC

00:14:29
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Stage	Management	Treatment
Till Stage 3A	medical management (Since no bowel perforation)	1. Nil Per Oral (NPO). 2. TPN (Total Parenteral Nutrition) : IV Fluids with Dextrose. 3. IV antibiotics : • Ampicillin (penicillin group). • Gentamycin (aminoglycosides). • Metronidazole (anaerobic coverage). Duration of Antibiotics : Stage 1A : 3 days. Stage 1B : 7–10 days. Stage 2A, 2B, 3A : 14 days.
Stage 3B	Surgical management.	• Stable → Laparotomy (Resection of necrotized part, followed by anastomoses). • Unstable → Perform PPD (Primary Peritoneal Drainage). When Stabilized, definitive surgical intervention.

Prevention of NEC :

1. Antenatal steroids : Dexamethasone/ Betamethasone.
   - They help in maturity of lungs → Preventing RDS (Respiratory Distress Syndrome).
   - They also help in maturity of GIT → Preventing bacterial colonization → Preventing NEC.
2. Minimal Enteral Nutrition (MEN) : Especially helpful in small, preterm babies.

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- In babies < 32 weeks of gestation or hemodynamically unstable preterm babies, no breast milk is given, only TPN is given.
- They should be given small amounts of enteral feeds (MEN) like expressed breast milk, using nasogastric tubes (10 ml/kg/day).
- This MEN helps in promoting the maturation of the GIT, decreasing the risk of NEC.
- AKA Trophic Feeds (Trophic = Growth).

3. Enterally fed probiotics.
- Normal gut flora like Lactobacillus/bifidobacterium spp.
- They prevent colonization from the pathogens.

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RESPIRATORY DISTRESS IN NEWBORN

Features of respiratory distress 00:00:24

Any 2 of the following must be present.
Respiratory rate > 60/min.
Chest retractions/indrawing.
Grunting & cyanosis : Grunting → Expiration in a partially closed glottis by the baby to prevent collapse of alveoli. Produces a positive end expiratory pressure that prevents alveolar collapse.

Causes of respiratory distress :

Pulmonary (more common)	Non pulmonary
1. TTNB : Transient Tachypnea of Newborn (m/c). 2. Respiratory distress syndrome (RDS). 3. Meconium Aspiration Syndrome (MAS). 4. Air leaks(Pneumothorax). 5. Pneumonia. 6. Anomalies : Congenital Diaphragmatic Hernia (CDH).	1. Cardiac failure due to certain congenital heart diseases. 2. metabolic : • Hypoglycemia. • Hypocalcemia.

monitoring of respiratory distress : 2 important scoring systems. 60c6b3eeaa8ded0e4e7e5ea7

1. Silverman Anderson Score.
2. Downe's Score.

Silverman Anderson Score : Useful for monitoring preterm babies with respiratory distress.

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Symptom	0	1	2
Upper chest retractions (Compare movements of chest with abdomen)	Synchronized (Chest = Abdomen)	Chest lags during breathing (abdomen moving outwards during inspiration, chest only slightly moving outwards)	See saw respiration (abdomen moving outwards, chest moving inwards : Severe)
Lower chest retractions	No	Mild	Marked
Nares dilatation/ nasal flaring	No	Mild	Marked
Xiphoid retraction	No	Mild	Marked
Grunting	No	Heard with stethoscope	Naked ear

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Interpretation :

Minimum score 0 : Normal.

Maximum score 10 : Completely abnormal.

Score	Degree of respiratory distress
< 5	Mild respiratory distress
5 – 7	Moderate respiratory distress
> 7	Severe respiratory distress. Impending respiratory failure that requires advanced forms of ventilatory support.

Active space

Downe's Vidyasagar score

00:10:04

Symptoms	0	1	2
Respiratory Rate	< 60/min	60 – 80/min	> 80/min or apnea
Cyanosis	No	In room air	With O_a support at $FiO_a \geq 40\%$
Air entry	Normal	Decreased	Barely audible
Retractions	No	Mild	Marked
Grunting	No	Heard with stethoscope	Naked ear

Interpretation :

>6 : Severe distress and impending respiratory failure.

Monitoring of oxygen saturation :

SpO_a <90% : Hypoxia.

Oxygen challenge : Give O_a and observe change in SpO_a.

≥ 20% improvement	No change or < 20%
Respiratory cause	Cardiac cause

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Transient Tachypnea of Newborn

00:15:10

Risk factors :

Lower segment Caesarean Section (LSCS) in term/ late preterm (35 – 36 weeks).

Diabetic mothers.

Reason : Delayed clearance of lung fluids.

Mechanisms :

In normal vaginal delivery, baby is squeezed & compressed and then delivered. The fluid is squeezed out of the lungs and baby can breathe freely.

But in LSCS, no squeezing causes remnant fluid in the lungs and leading to distress.

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During delivery : Increased expression of ENaC & Na^+ K^+ ATPase channels. Helps in sodium and water reabsorption, clearing lung fluid. In LSCS, impaired expression of these channels.

Presentation : Breathing difficulty within 6 hours after birth.
Chest X ray : Fluid in the lungs.
Perihilar streaks/sun burst appearance : Fluid in the bronchopulmonary structures.
Fluid in the interlobar fissure.

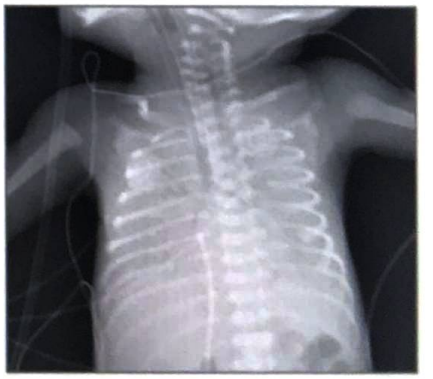
Management :
Transient condition lasts upto 72 hours.
So only supportive treatment with O_2 inhalation.

New guidelines of TTNB :
Salbutamol inhalation in early TTNB increases expression of ENaC and Na^+ K^+ ATPase channels.

Respiratory distress syndrome

00:21:25

Occurs in preterm (<35 weeks) babies. Surfactants become mature after 35 weeks.



Pathology : Hyaline Membrane Disease (HMD).

Reason : Deficit or immature surfactant levels.

Surfactant :

- Produced by Type 2 pneumocytes.
- Keeps the alveoli open by decreasing the surface tension and prevents its collapse.

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In RDS, Immature/deficit surfactant : Collapsed alveoli → hypoxia and respiratory distress.
Presentation : < 6 hours.

Active space

Chest X Ray : White out lungs/ground glass appearance (also seen in PAP).
Air bronchogram sign : Air enters the patent bronchi and appears black.

Pulmonary Alveolar Proteinosis (PAP) : Presents with White out lungs/ground glass appearance similar to RDS. Autosomal recessive fatal condition.
Reason : Accumulation of old ineffective surfactant.
Normally, old surfactant is removed by alveolar macrophages.
In PAP, surfactant is not degraded and accumulates.
No functional surfactant.

Tests for fetal lung maturity 00:27:55

These can be done in amniotic fluid/ gastric juice after birth but are obsolete.

- Lecithin : Sphingomyelin ratio (L : S) : Normally > 2 : 1.
  Phospholipids that compose surfactant. Lecithin is the mature form, sphingomyelin is the immature form. more lecithin, more mature surfactant.
  Exception : Infant of diabetic mother (IDM) :
  should be > 3.5 : 1.

- Phosphatidyl glycerol test : Better and more sensitive.

- Lamellar body count : Storage form of surfactant in Type 2 pneumocytes. Some detected in amniotic fluid.
  Mature lung : > 50,000/microlitre.

- Shake test : 0.5 ml of gastric aspirate + 0.5 ml 95% ethyl alcohol. Shake it vigorously for 10 – 15 min and leave it for another 10 – 15 min. Full rim of bubbles indicate positive test for lung maturation.

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Management of Respiratory Distress Syndrome

00:31:52

Delivery Room :

Preterm baby <34 - 35 weeks with breathing difficulty.

Suspect RDS.

Start CPAP (Continuous Positive Airway Pressure) :

- Keeps alveoli open & prevents its collapse.

< 28 weeks of gestation : Prophylactic surfactant to prevent RDS.

This strategy is termed as early delivery room cpap with selective surfactant.

ICU : Depends on Silverman scoring.

RDS	Management
< 5 : Mild RDS.	Start warm humidified O_a. Target SpO_a levels : 90 - 95%. SpO2 > 95% : Excess O2 → Retinopathy of prematurity leading to blindness.
5 - 7 : Moderate RDS.	Start CPAP. Initial Pressure : 5cm H_aO. Pressure and O_a concentration can be adjusted according to the response in the baby.
> 7 : Severe RDS.	Start mechanical ventilation + endotracheal surfactant therapy. Standard guideline for surfactant therapy : INSURE technique. Intubation → Surfactant → Extubation. (Standard Guideline)

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Newer techniques for surfactant therapy 00:38:26

Non invasive/less invasive.

MIST : Minimally Invasive Surfactant therapy.
LISA : Less Invasive Surfactant Administration.

} Done using a feeding tube/ tracheal catheter

Early rescue surfactant therapy :

Moderate RDS
↓
CPAP
↓
No response (FiO2 >40%)
↓
Surfactant therapy
↓
Decrease the need for mechanical ventilation

Indications for surfactant therapy :

- Severe RDS.
- Moderate RDS with no response to CPAP.

Antenatal prevention of RDS :

Corticosteroids : To mothers who deliver at 24 – 34 weeks.

Corticosteroids	Dose
Betamethasone	12 mg/dose, 2 doses 24 hours apart
Dexamethasone (common in India)	6 mg/dose, 4 doses 12 hours apart

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Benefits : Decreases incidence of

- Respiratory distress syndrome.
- Necrotizing enterocolitis.
- Intra ventricular hemorrhage in preterm babies.
- Overall neonatal mortality.

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Bronchopulmonary dysplasia(BPD)/ Chronic Lung Disease of newborn

00:44:31

Risk factors :

Preterm babies born at < 32 weeks of gestation.

Babies on O_a support for > 4 weeks after birth.

When to assess ? 36 weeks of post menstrual age.

BPD	O_a requirement
Mild BPD	No O_a required
Moderate BPD	FiO_a < 30%
Severe BPD	FiO_a > 30% or any form of ventilatory assistance

Congenital diaphragmatic hernia :

Intestines herniating from the abdominal cavity into the thoracic cavity.

Type 3eeaa8ded0e4e7e5ea7

Type of hernia	Location of defect
Bochdalek hernia (m/c type)	Posterolateral and left side of diaphragm
Morgagni hernia (rare)	Anterior aspect and right side of diaphragm

Features of CDH :

Compression of lungs → Lung hypoplasia → Respiratory distress after birth.

Heart sounds on the right side due to right shift of the thoracic contents.

Sunken (scaphoid) abdomen.

Active space

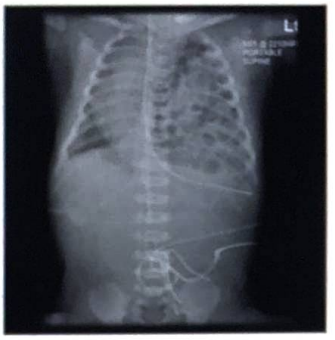
Diagnosis of CDH

00:51:30

Antenatal : USG between 16 - 24 weeks.

After birth : Chest X ray.

- Air shadows in thoracic cavity indicating intestinal shadows.
- No definite diaphragm border on the left indicating defect in the diaphragm.
- Heart shadow on the right side.
- Absent air shadows in the abdomen.



Management :

Medical Management	Surgical Management
First 48 hours after birth	> 48 hours of life
Aim : • Promote lung expansion. • Stabilize pulmonary hypertension due to lung compression. Treatment : Ventilation Conventional Ventilation. ↓ HFOV (High Frequency Oscillatory ventilation). ↓ ECMO (Extra Corporeal Membrane Oxygenation).	Treatment : • Native tissue repair. • Patch Repair (Polytetrafluoroethylene : GORE- TEX). Approach : Subcostal approach as it helps in better visualization.

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Poor prognostic factors of CDH :

Antenatal factors	Postnatal factors
USG : • Lung head ratio < 1. • Liver in thoracic cavity. MRI : • Derived Total Lung volume (TLV) < 20 ml.	• Degree of pulmonary hypertension : most important prognostic factor. • Size of the defect. • Early onset of respiratory distress <24 hours. • Requires ECMO.

Meconium Aspiration Syndrome 00:01:03

Common in post term babies (or) term babies with IUGR :

Intrauterine stress

↓

Increased motilin

↓

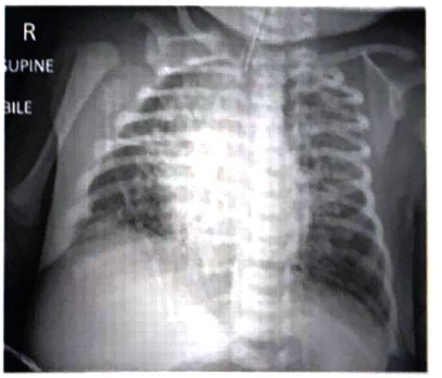
Increased peristalsis of GIT

↓

Expulsion of meconium in utero into amniotic cavity

↓

Swallowed and aspirated → MAS



Chemical Pneumonitis

Consequences of MAS :

Partial block of a main airway : Obstructive emphysema → Air trapping during exhalation → Hyperinflation in CXR.

Complete block of a small airway : Segmental atelectasis.

Meconium in alveoli : Chemical pneumonitis.

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Diagnostic criteria for MAS 01:08:29

Diagnosis of exclusion :

- Meconium Stained Liquor (MSL) at birth + Respiratory distress.
- X ray : Coarse, ill defined infiltrates.
- Exclude other Known conditions.

Complication of MAS :

- Air leaks like Pneumothorax.
- Persistent Pulmonary Hypertension of Newborn (PPHN).

Management :

- Symptomatic management according to ventilatory requirements like O_a therapy or CPAP.
- PPHN : Inhaled Nitric Oxide (iNO).

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NEONATAL HYPOGLYCEMIA

Hypoglycemia definition (WHO) : Blood glucose < 45 mg/dl.
Hypoglycemia can also be asymptomatic.

Screening for neonatal hypoglycemia 00:01:16

'At risk' babies :

- Low substrate (low glycogen) : Preterm babies < 35 weeks of gestation, low birth weight <2 kg, intrauterine growth restriction (IUGR).
- Relative hyperinsulinemia : Large for gestation babies, infant of diabetic mother, Rh incompatibility (transient insulin increase)
- Sick baby : Neonatal sepsis, birth asphyxia, hypothermia.

Hypoglycemia is screened by dipstick measurement of heel prick sample.

Schedule of screening for at risk babies : 60c6b3eeaa8ded0e4e7e5ea7
At 2, 6, 12, 24, 48 and 72 hours after birth.

Features of neonatal hypoglycemia :

- Earliest : Jitteriness / tremors.
- Other features : Lethargy, weak cry, poor feeding pattern.
- Can present with seizures.
- Autonomic changes include sudden changes in heart rate, sudden pallor or episodes of hypothermia (rare in a newborn compared to older child or adults).

Jitteriness vs seizures :

	Jitteriness	Seizures
Stimulus	Stimulus sensitive	Not stimulus sensitive
Eye deviation	Not seen	Seen

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Autonomic changes like sudden change in heart rate, respiratory pattern	Not seen	Seen

Management :

Depends on symptomatic or asymptomatic.

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Symptomatic hypoglycemia :

- Emergency : Treated immediately with i.v. bolus 10% Dextrose (2 ml/kg) followed by i.v. 10% Dextrose infusion at 6 mg/kg/min.

Asymptomatic hypoglycemia (detected while screening) :

- < 20 mg/dl : i.v 10% dextrose infusion at 6 mg/kg/min.
- 20 – 45 mg/dl : Oral feeds.
  Feed once → Recheck after 30 to 45 minutes.
  If normal → continue breastfeeding every 2 hourly or as per baby's requirements.
  If <45 mg/dl → Start i.v 10% dextrose infusion.

Infant of diabetic mother

00:10:36

Pedersen's hypothesis : maternal hyperglycemia → sugar crosses placenta → hyperglycemia in fetus → increased secretion of insulin in fetus → excess growth (anabolism) → macrosomia (large for gestational age).

Complications of infant of diabetic mother :

- Delivery complications : Shoulder dystocia, brachial plexus injury i.e. Erb's palsy, fractures of clavicle.
- Asymmetrical interventricular septal hypertrophy (form of hypertrophic cardiomyopathy) : Transient condition.
- Increased demand of oxygen → increased RBC production → polycythemia → predisposition to thromboembolic manifestations (hypercoagulable blood)
- Increased insulin → interferes with surfactant maturation → respiratory distress syndrome.

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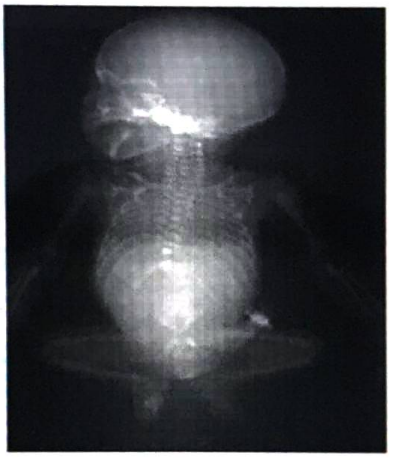
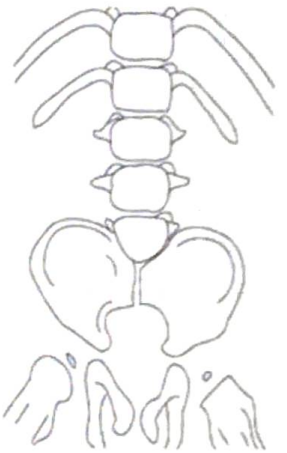
- Increased insulin → Impaired gut motility (colon) → lazy left colon syndrome → can present with delayed passage of meconium.
- Increased sugar in the fetus → congenital anomalies.
  m/c system affected : Cardiovascular system.
  m/c heart defect : Ventricular septal defect.
  m/c group of anomalies : Neural tube defect.
  most specific heart defect : Transposition of great arteries.
  Overall most specific anomaly : Caudal regression syndrome.
  Example : Sacral agenesis.





Single most common anomaly in an infant born to diabetic mother is VSD.

- After birth of baby : Hypoglycemia, hypocalcemia, hypomagnesemia.

Most common metabolic abnormality associated with seizure in a baby born to diabetic mother :
Hypoglycemia > Hypocalcemia.

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NEONATAL JAUNDICE

Bilirubin metabolism

00:00:40

Jaundice : Increased levels of bilirubin manifesting as yellowish discoloration.

RBC

↓

Heme

↓ Heme Oxygenase

Biliverdin

↓ Bilurubin reductase

Unconjugated bilirubin (water insoluble, cannot be excreted)

↓

UDP glucuronide transferase (UDP-GT) in liver

↓

Conjugated bilirubin (water soluble, can be excreted)

↓

Bile in bile duct

↓

2nd part of duodenum

↓

Intestine : Colonic bacteria

↓

90% stercobilinogen (yellow stools)

<10% urobilinogen (light yellow urine)

Very small portion taken back to liver : Enterohepatic circulation

Assessment of jaundice :

Serum bilirubin :

Bilirubin	Normal levels
Total	1 – 1.3 mg/ dL
Direct (conjugated)	≤ 0.3 mg/ dL
Indirect (unconjugated)	0.9 mg/dL

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Visible jaundice : ≥ 5 mg/dL.

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Screening tests for jaundice

00:06:17

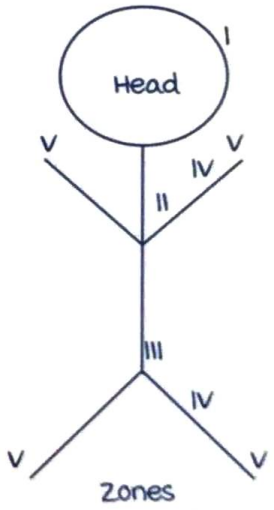
1. Transcutaneous bilirubinometer (TCB) : Hand held device.
   Sternum : Common site.
   Useful only if baby is > 35 weeks of gestation.
   Reliable only if baby is > 24 hours old.

2. Visual assessment by Kramer's rules :
   States that jaundice spreads in cephalo caudal progression from head to trunk and limbs.
   1st develops in the eyes.

Zones of jaundice :

Zones	Part of the body	Bilirubin level
I	Head : Eyes	6 mg/ dL
II	Trunk	9 mg/ dL
III	Lower abdomen & thigh	12 mg/dL
IV	Upper & lower limbs	15 mg/dL
V	Palms & soles	>15 mg/ dL

Unconjugated bilirubin(>15 mg/dl) : Water insoluble,
Fat soluble
↓
Cross blood brain barrier
↓
Brain damage



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Forms of jaundice

00:12:01

MC : Physiological >> pathological.

Physiological jaundice :

Increase in unconjugated bilirubin always.

Due to immature conjugating enzyme : UDP glucuronyl transferase.

Appears by 24 to 72 hours of age.

Peaks by day 3.

May present upto 1 – 2 weeks.

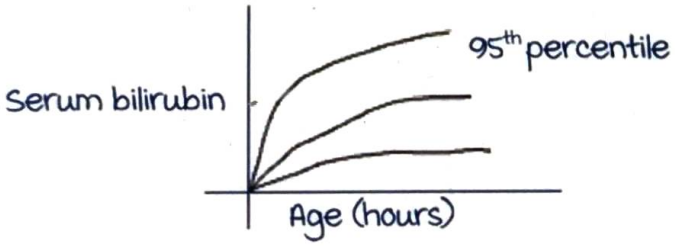
No treatment required.

Pathological jaundice :

- Day 1 : Any jaundice (> 5 mg/dL). But, if it appears between 18 – 24 hours after birth, could be physiological.
- Day 2 : Jaundice in arms and legs (> 10 mg/dL).
- Thereafter jaundice > 12 to 14 mg/dl is pathological
- Anytime : Jaundice in palms & soles (> 15 mg /dL).
- Rate of rise in bilirubin >5 mg/dl /day or > 0.2 mg/dL/hr.
- TSB nomograms : Above 95th percentiles for age.
- Signs of bilirubin encephalopathy.
- Any jaundice persisting > 2 weeks in term babies and > 3 weeks in preterm babies.

Total Serum Bilirubin (TSB) nomograms :

Compare serum bilirubin with age of the baby in hours after birth.



These percentile lines depend on gestational age, risk factors like asphyxia, sepsis, hypothermia.

Any value > 95th percentile is significant and pathological and indicates that the jaundice requires treatment by phototherapy or exchange transfusion.

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Etiology of pathological jaundice

00:20:33

Etiology	Conditions
Hemolysis	Inherited : G6PD, hereditary spherocytosis. Acquired : maternal antibodies mediated lysis of baby's RBCs (Rh/ ABO blood group incompatibility).
UDP-GT defects	Criggler Najjar syndrome (AR) : Type 1 : Absent UDP-GT. Type 2 : Low levels of UDP-GT. Hypothyroidism Sepsis.
Increased entero hepatic circulation	Breast feeding issues. Intestinal obstruction.
Extravasated blood	Cephalohematoma, bruising.
Increased conjugated bilirubin	Neonatal cholestasis.

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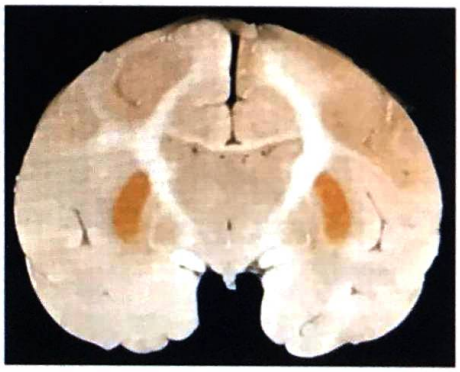
Complication :

Bilirubin Encephalopathy/BIND (Bilirubin Induced Neurological Damage) :

- Brain damage due to increased levels of unconjugated bilirubin as it is lipid soluble and can cross blood brain barrier.
- Area affected : Basal ganglia.
- Pathologically, called Kernicterus.
- May be acute or chronic.



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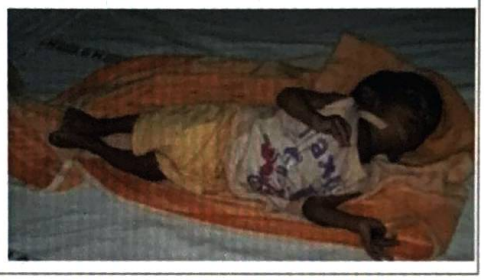
Acute	Chronic
0 – 3 days : • Hypotonia. • Decreased activity. • High pitch (shrill) cry. >3 days : • Hypertonia. • Seizures. • Fever.	Older children. Basal ganglia damage : • Extrapyramidal Cerebral Palsy (CP) : Chorea, athetosis, abnormal tone, posture etc., • Sensorineural hearing loss (SNHL). • Upward gaze palsy. • Dental problems : Enamel dysplasia.
Acute BIND : Hypertonia Opisthotonos. Retrocollis : Hyperextended neck.	

Management of neonatal jaundice 00:30:05

Monitor : Visual assessment ± TCB every 8 – 12 hours for 3 – 5 days after birth.

Step 1 : Look for serious jaundice. Requires immediate treatment.
- Any jaundice on day 1.
- Jaundice in palms & soles.
- TSB nomogram : > 95th percentile for age.
- Signs of bilirubin encephalopathy.

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Step 2 : Look for signs of bilirubin encephalopathy.

Present	Absent
Exchange transfusion.	Phototherapy (PT).

Active space

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Send serum bilirubin for further management.

↓

If, no features of serious jaundice.

↓

Step 3 : Does baby have significant jaundice ?

Day 2 : Jaundice in arms/ legs.

Rate of rise in bilirubin : > 5 mg/ dL/day or > 0.2 mg/dL/hr.

Jaundice persists for >2 weeks in term babies & >3 weeks in preterm babies.

Significant Jaundice

↓ Present ↓ Absent

Investigations :

Serum bilirubin in Normogram.

↓ Value above cut off in Normogram : Start PT.

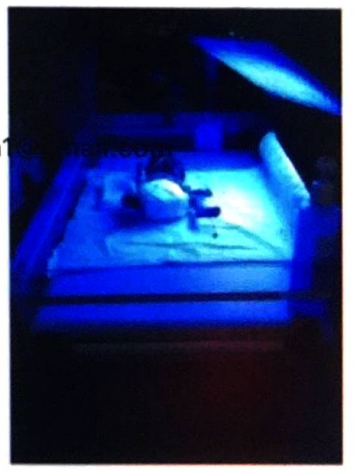
↓ Value below cutoff → Continue Observation.

Treatment of neonatal jaundice

00:36:45

Phototherapy :

UV Rays : 460 – 490 nm.



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Lamps :

Compact Fluorescent Lamps (CFL).

Light Emitting Diode (LED).

Usage : CFL > LED.

Efficacy : LED > CFL.

Distance between baby and light : 30 – 45 cm.

Baby must be naked except for eyes and gonads as it can cause damage to retina and testis.

Effective irradiance : At least 30 $\mu W/cm^2/nm$.

Active space

Complications :

- Damage to retina & testis. Avoided by covering them.
- Dehydration due to heat. Avoided by increased duration & frequency of breast feeding.
- Hypocalcemia.
- Bronze baby syndrome : Occurs in baby with increased conjugated bilirubin or in baby with hepatic dysfunction. Formation of brown pigmentation all over baby's skin and in body fluids. Its an avoidable complication as phototherapy should not be given in babies with Conjugated bilurubinemia

Exchange Transfusion (ET)/DVET :

Indications :

- Signs of bilirubin encephalopathy.
- Rh incompatibility : Cord blood bilirubin > 5 mg/dL or hemoglobin < 10 mg/dL → Significant hemolysis.
- Nomograms : Cut off values > ET range → Start ET.

Procedure of ET

00:42:26

Exchange of baby's blood with twice its volume of fresh blood. Hence, also called Double Volume Exchange Transfusion (DVET).

Normal volume : 80 – 90 ml/kg.

Volume transfused : 6006b3ea8ded0e4e7e5ea7

Type of blood :

Cross matched with baby's blood as well as mother's blood.

O -ve blood commonly used as it doesn't contain any antigens.

Condition	Blood type
Rh Incompatibility	Rh -ve & blood group 'O' suspended in AB plasma.
ABO incompatibility	Rh compatible with baby's blood & blood group 'O' suspended in AB plasma.
Other indications	Rh & blood group compatible with baby.

Active space

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Prolonged Jaundice in Newborn :

Persistent Jaundice > 2 weeks in term,

> 3 weeks in preterm.

Risk factors :

Common	Uncommon
Breast feeding issues : Transient conditions. 1. Breast feeding Jaundice : Decreased duration/ frequency of feeds. 2. Breast milk Jaundice : Milk contains components (e.g., Pregnanediol) that interfere with conjugation. Treatment : Continue breast feeds.	Inherited disorders : Criggler Najjar syndrome.
Continuing hemolysis.	Hypothyroidism.
Cholestasis.	Extravasated blood.

Management of prolonged jaundice 00:50:03

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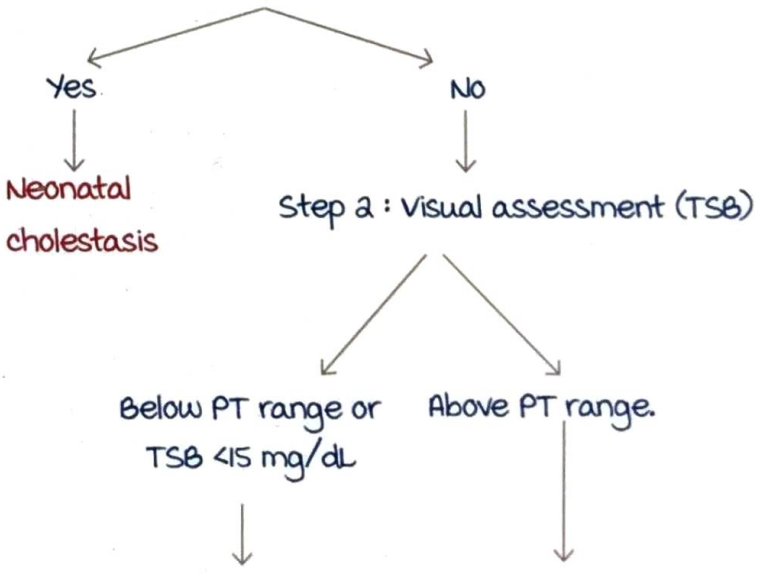
Step 1 : Does baby have features of cholestasis ?



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Continue observation.

MC cause : Breast feeding issues.

Start PT.

Evaluate for causes.

i) G6PD.

ii) Rh / ABO incompatibility.

iii) Thyroid function test

↓ (-)

Crigler Najjar syndrome

Type I : Severe, may require liver transplantation.

Type 2 : Phenobarbitone, induces UDP GT.

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NORMAL GROWTH

Growth phases 00:00:16

Periods of growth :

1. Prenatal period : Growth before birth/intrauterine period.
   - Fertilized ovum/zygote : First 2 weeks of gestation.
   - Embryo : 2 to 8 weeks of gestation.
   - Fetus : 9 weeks of gestation.
2. Postnatal period :
   - Newborn : First 4 weeks.
     Early newborn : First week.
     Late newborn : Next 3 weeks.
   - Infancy : First year.
   - Toddler : 1 to 3 years.
   - Preschool : 3 to 6 years.
   - School age : 6 to 12 years.
   - Adolescent : 10 to 19 years.

Adolescent age group :

Early : 10 to 13 years.
Mid : 14 to 16 years.
Late : 17 to 19 years.

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Pubertal changes :

Puberty occurs earlier in females (8 to 13 years) than males (9 to 14 years).

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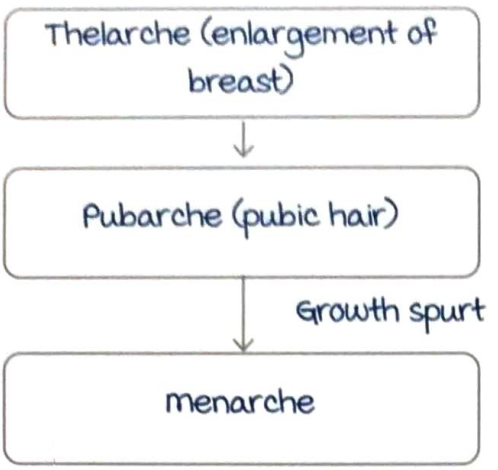
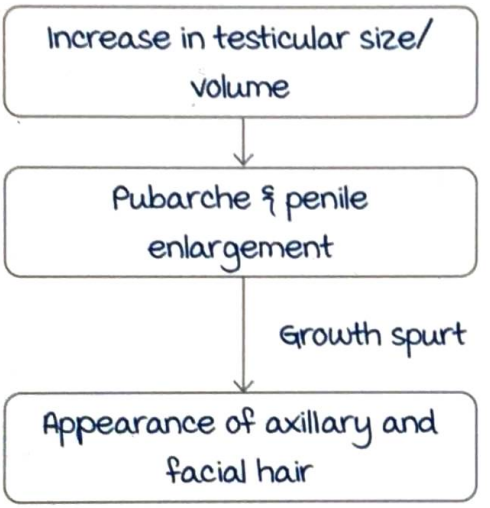
Sequence in females :



Sequence in males :

Assessment of testicular size is by Praders orchidometer.



Changes during the middle part of puberty in males :

- Spermarche (production of sperms).
- Voice cracking/deepening seen.

Assessment of puberty :

Tanner's SMR (Sexual maturity rating).

Done separately in males and females.

In males : Testis, penis, pubic hair, and scrotum.

In females : Breast and pubic hair appearance.

From stages I (prepubertal) to stages V (adult like appearance).

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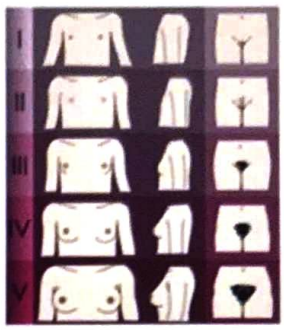
Tanner's staging in females : Breast appearance.



Stage I : No changes in the breast.

Stage 2 : Appearance/prominence of breast buds.

Stage 3 : Generalized swelling of the breasts (beyond nipple and areola).

Stage 4 : Nipple + areola forming a second mount over the breast.

Stage 5 : Mature/adult type appearance, where the nipple protrudes forward, and areola retracts.

Pubic hair appearance :

Stage I : No pubic hairs.

Stage II : Straight, sparse hairs.

Stage III : Increase in number and curling of hairs.

Stage IV : Dense pubic hair (curly).

Pigmentation starts increasing from stage II to IV.

Stage V : Pubic hairs extends into medial part of thigh region.

Tanner's staging in males :

Pubic hair changes are similar to females.

Testicular volume is represented as number.

Size of the testis is mentioned.

Stage I : Testis size < 2.5 cm and volume < 4 ml.

Stage II : Testis size ≥ 2.5 cm and volume > 4 ml.

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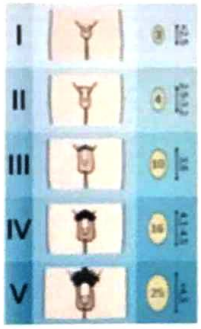
Scrotum size increases, skin over scrotum is red in color.



Stage III : Further increase in volume of testis, penis lengthens, and scrotal skin is dark.

Stage IV : Increase in length and breadth of the penis.

Stage V : Volume of testis > 20 ml (adult like appearance).

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Growth patterns

00:15:10

Rules :

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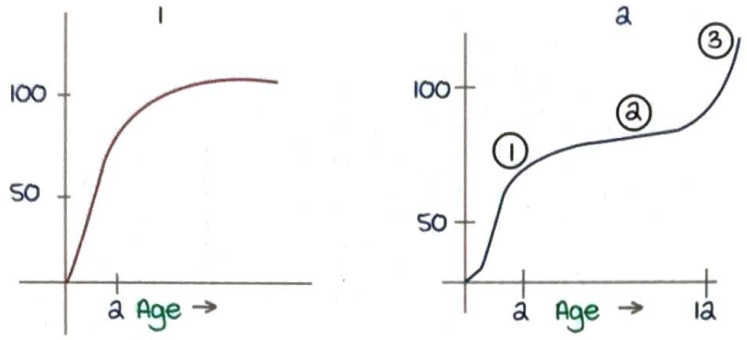
Growth is in Cephalocaudal progression.

Growth occurs from the distal to proximal direction.

Not all tissues in the body grow at the same rate.



Graph 1 : Till 2 years there is steep increase followed by minimal growth (parabola). Depicts growth of brain.

Graph 2 : Depicts somatic growth.

Sigmoid growth.

Phases : Rapid growth (1 & 3) : Growth spurts.

Phase 3 : Pubertal growth spurt.

Pubertal growth spurts occur early in females.

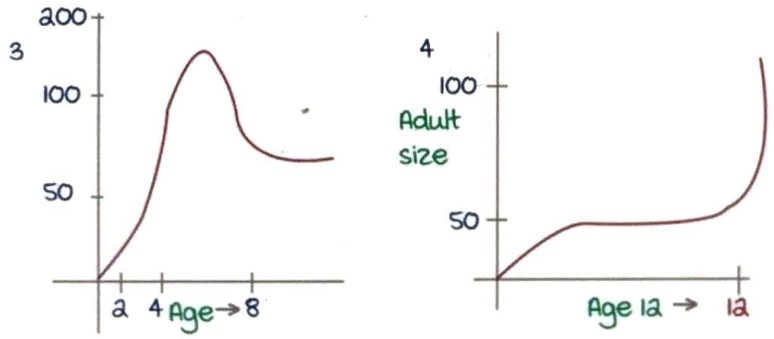
Increase in height is around 16 to 28 cm in females.

Increase in height is 20 to 30 cm in males.

Growth spurts :

Female : Tanner's stage 3.

Males : Tanner's stage 4.



Graph 3 : Peak (4–8 years) is 2 times of adult size. Come back to normal size after 8 years.

Eg : Lymphoid tissues like LN, tonsils, and adenoids.

Active space

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Between 4 to 8 years : Physiological lymphoid hyperplasia.

Graph 4 : Increase in size after 10 to 12 years.
Stage of gonadal growth.

Assessment of growth

00:23:34

Anthropometry : Measurement of growth parameters.
Average birth weight in India : 2.9 Kg.
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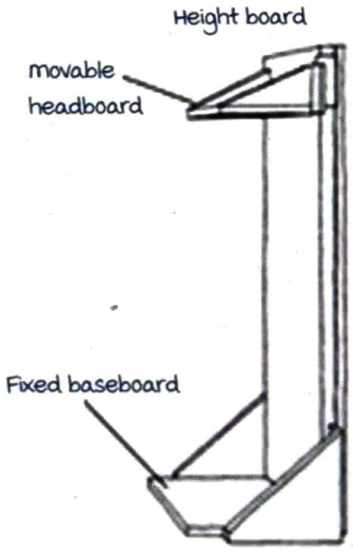
LBW is defined as weight < 2.5 Kg.
During first week, there is 10% of weight loss in term babies.
For preterm babies : Up to 15% of weight loss.
This is called as physiological weight loss.

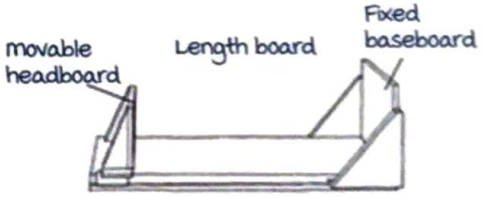
Regaining of birth weight : 10 (term) to 14 days (preterm).
Best way of assessment : Growth charts.

Birth weight doubles by 5 to 6 months.
Birth weight triples by 1 year.
Birth weight quadruples by 2 years.
Height/length assessment :
Length : measured in supine posture (up to 2 years).
Measured by stadiometer (height) or infantometer (length).
Staiometer



Infantometer

move quickly and surely to measure length accurately before the baby becomes agitated.



Normal length at birth : Approximately 50 cm.
In the first year : Increase of 25 cm. So, at 1 year : 75 cm.
In the second year : Increase of 10 to 15 cm. So, at 2 years :

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85 to 90 cm.
There is a steady increase of 6 cm/year till 12 years.
After 12 years (pubertal growth spurts) :
Females : 8cm/year.
Males : 10 cm/year.

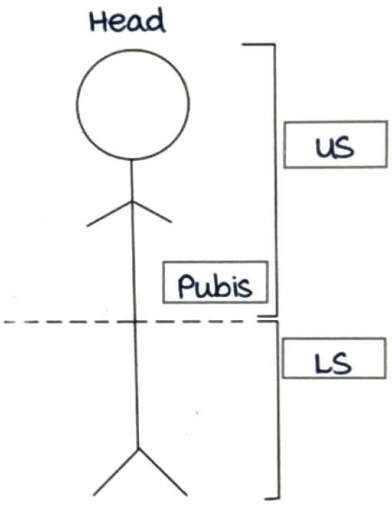
Height doubles by 4 years.
Height triples by 12 years.

Half of adult's height in a normal child (80–85 cm) : Around 2 years (18 to 24 months).

Body proportions

00:32:28

Upper Segment (US): Lower Segment (LS).
At birth it is 1.7 : 1
At 3 years it is 1.3 : 1
At 7 to 10 years it is 1 : 1.
After >10 years it is 0.9 : 1.



They are useful in the evaluation of short stature.
In tall children : Arm span.

At birth : Height > Arm span (difference is 2.5 cm).
At 11 years : Height = Arm span.
>11 years : Arm span > Height (difference is 1 to 2 cm).

Circumferences

00:36:50

Head circumference (HC) : Also called as occipitofrontal circumference.
It indicates brain growth.

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At birth : Approximately 34 cm (33–35 cm).
First 3 months after birth : Increase of 2 cm/month.
At 3 months : 40 cm.

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Next 3 months : Increase in 1 cm/month. At 6 months : 43 cm.
Next 6 months : Increase in 0.5 cm/month. At 12 months : 46 cm.
Next 1 year : Increase in 2 cm. At 2 years : 48 cm.
At around 12 years : 52 cm.

Chest circumference (CC) : measured at the level of nipples midway between inspiration and expiration.

Chest circumference

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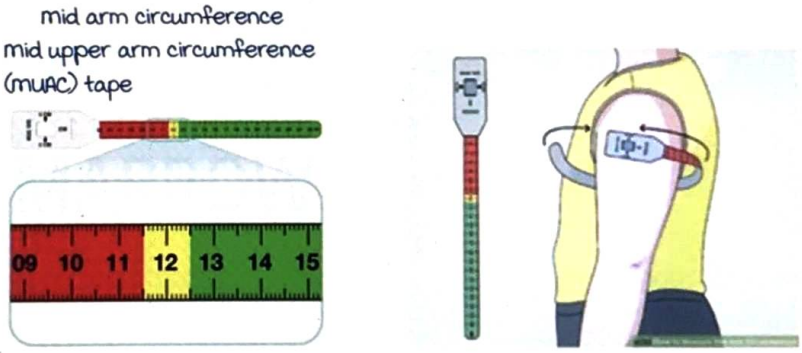
Birth : HC > CC (3cm).
1 year : HC = CC.
> 1 year : CC>HC.

Mid arm circumference :



measured in children < 5 years.
It is useful for community assessment of growth, because it can be done by health workers.

Shakir's tape :
Midpoint of the arm : Mid point between 2 bony prominences.
Green : Nutrition is normal.
Yellow : Malnutrition.
Red : Severe malnutrition (≤ 11.5 cm).

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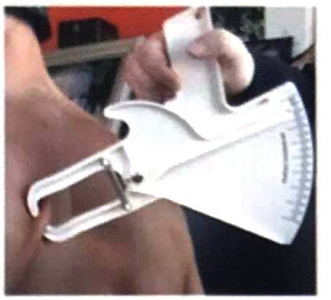
Skin fold thickness measurement :

Thickness of subcutaneous fat is measured.

It is an indicator of nutritional status in children.

It is done by Harpenden's caliper.



≥ 10 mm : Normal.

< 6 mm : Severe malnutrition.

Charts for skin fold thickness : Interpreted as percentile value.

< 5th percentile : Low value (malnutrition).

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Growth charts

00:46:06

WHO growth is most preferred and used commonly.

In India, between 0 to 5 years : Only WHO charts are used.

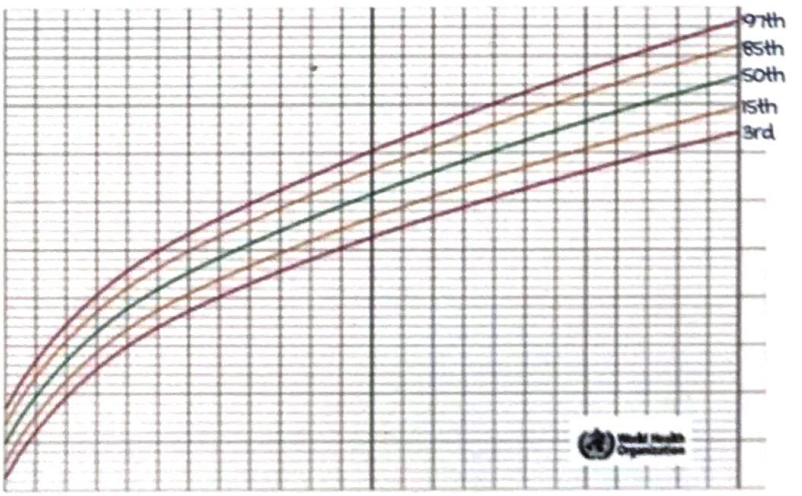
5 to 19 years : WHO or IAP charts can be used.

WHO charts can be used as international comparison tools.

Types of WHO charts :

- Weight/age charts.
- Weight/height charts.
- Height/age charts.
- HC for age charts.
- MAC for age charts.
- BMI charts (weight in Kg/height in m^2) : Used in > 5 years of age.

Best tool for assessment in < 5 years : Weight /Height chart.



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Growth chart in

Blue colour : Boys.

Pink colour : Girls.

In a height for age chart,

Major percentiles are recorded.

50th percentile : mean value.

High value (tall stature) : Value above 97th percentile.

Low value (short stature) : Below 3rd percentile.

Between 97th percentile and 3rd percentile : Normal values.

Standard deviation /Z scores : Deviation from mean value.

Eg : 97th percentile (+2) SD

85th percentile is (+ 1) SD.

15th percentile is (- 1) SD.

3rd percentile is (- 2) SD.

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ABNORMALITIES IN HEAD SIZE AND HEAD SHAPE

Microcephaly (small head) :

Based on head circumference for age chart, head circumference < -3SD is considered as microcephaly.

This is an exception to general rule : Considering < -2SD in the definition would mean including normal children as well.

Primary/Genetic microcephaly :

Seen due to any insult during brain development.

Refers to anomalies affecting the brain like anencephaly or neuronal migration disorders.

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Genetic syndromes like down's Syndrome (trisomy 21).

Familial microcephaly : Small head size runs in the family members. Usually inherited as autosomal recessive.

Secondary microcephaly (normal brain development) :

Acquired microcephaly : Extrinsic factors cause microcephaly.

Extrinsic factors include :

- Maternal factors :
    1. Infections during intra uterine life.
       Example : TORCH infection.
    2. Radiation during 1st trimester (impaired organogensis).
    3. Toxins (drugs/chemicals) taken durig pregnancy.
    4. Teratogens like phenytoin or alcohol.
- Perinatal factor like birth asphyxia.
- Post natal factors :

  Trauma or infection in the child up to the age of 2 years (active brain growth occurs till 2 years of age).
- Metabolic disorders (metabolite accumulation in brain) :
    1. Phenylketonuria.
    2. Lysosomal storage disorders :
       Hepatosplenomegaly and low IQ are seen.
       a. Gaucher disease.
       b. Niemann pick disease.

       Both have autosomal recessive inheritance.

Active space

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Gaucher's disease	Niemann pick disease
Deficiency in glucocerebrosidase enzyme	Deficiency of sphingomyelinase
Can also affect the bones : Causes expansile lytic lesions also called as Erlenmeyer flask deformity and pancytopenia (defective bone marrow).	Does not involve the bones
No cherry red spots.	Cherry red spots seen in macula of eye.
Accumulation of glucocerebrosides as typical cytoplasmic inclusions called wrinkled/crumpled tissue paper appearance on biopsy.	Accumulation of sphingomyelin gives foamy appearance of cells.

Late onset syndromes associated with microcephaly :

- Rett syndrome (acquired microcephaly).
- Angelman Syndrome.
- Seckel's Syndrome.

Rett syndrome

00:12:56

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X-linked dominant disorder.

- Genetic disorder characterized by defect in MECP2 gene.
- From birth till 1 year of age, head circumference is normal. After 1 year, deceleration of brain growth results in microcephaly.
- Developmental regression :
  Up to 1 year of age, the child achieves normal milestones but after 1 year of age, the child loses previously acquired milestones (eg : stand without support in 1 year but not able to, even with support at 1.5 years).
- Stereotypies :
  Characteristic hand wringing
  purposeless repetitive
  movements in the midline.
- Speech defects.

Active space

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- Ataxia (worsen over time and adolescents become wheelchair-dependent.

Stereotypies

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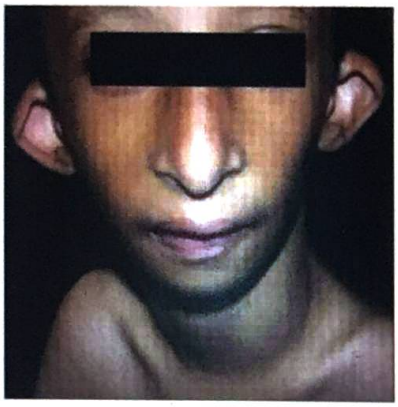
Seckel syndrome :

- Bird headed dwarfism.
- Beak like nose.



Seckel syndrome

Q. A child with microcephaly, low IQ, seizures and hypopigmented hair is being evaluated. The child is born out of consanguinous marriage. Which of the following is the likely cause of microcephaly?

- Familial microcephaly.
- Inborn error of metabolism.
- TORCH infections.
- Neural tube defect.

Diagnosis : Phenyl Ketonuria.

Phenyl Ketonuria :

Most common amino acid metabolic disorder.

Autosomal recessive in inheritance.

Normally, phenylalanine $\xrightarrow[\text{hydroxylase}]{\text{Phenylalanine}}$ Tyrosine $\longrightarrow$ melanin

Phenylketonuria $\longrightarrow$ deficient/absent phenylamine hydroxylase $\longrightarrow$ deficient tyrosine $\longrightarrow$ deficient melanin $\longrightarrow$ hypopigmentation $\longrightarrow$ fair skin, blonde hair, blue eyes.

Increasing phenylalanine $\longrightarrow$ converted to metabolites like phenylactate, phenylpyruvate & phenylacetate.

Brain damage and microcephaly in these patients.

Active space

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Musty/mousy odor of the urine → due to phenylacetate excretion.

↓

Management :

- Restrict diet containing phenylalanine.
- Enhance activity of phenylalanine enzyme co-factor, via synthetic BH4 (tetrahydrobiopterin) called as sapropterin.

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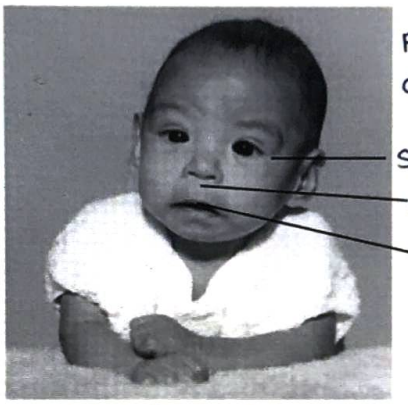
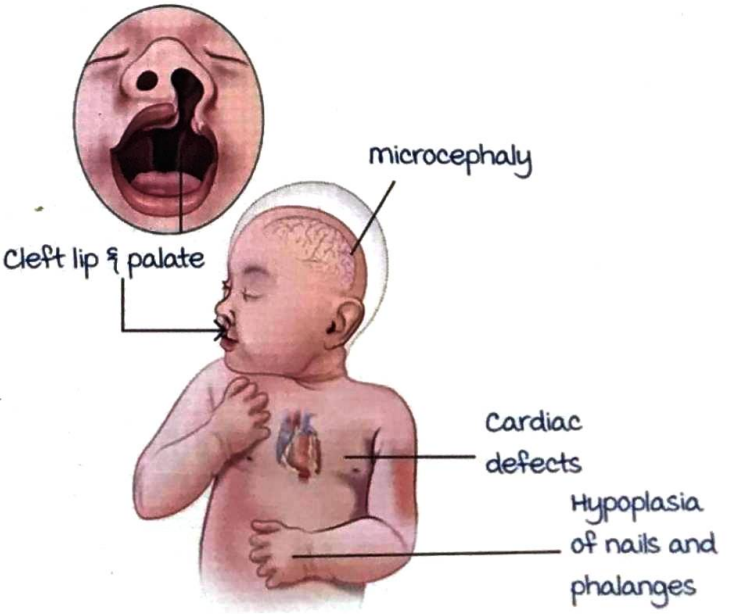
Q. Phenotypic appearance of two infants with microcephaly is shown below. Identify the possible teratogen incriminated in these scenarios ?



Fetal alcohol syndrome

Philtrum : Area between nose & mouth. Normally, markings/ impressions are seen.

In fetal alcohol syndrome, there is increased risk of heart defects (Ventricular Septal Defect is common).



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Fetal hydantoin syndrome :

- Phenytoin (diphenylhydantoin) exposure.
- Cleft lip/palate.
- Heart defects (commonly VSD).
- Small hands/feet.
- Mid facial hypoplasia (mid face structures smaller when compared to other structures in face).

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Macrocephaly :

Head circumference > +2SD for the age.

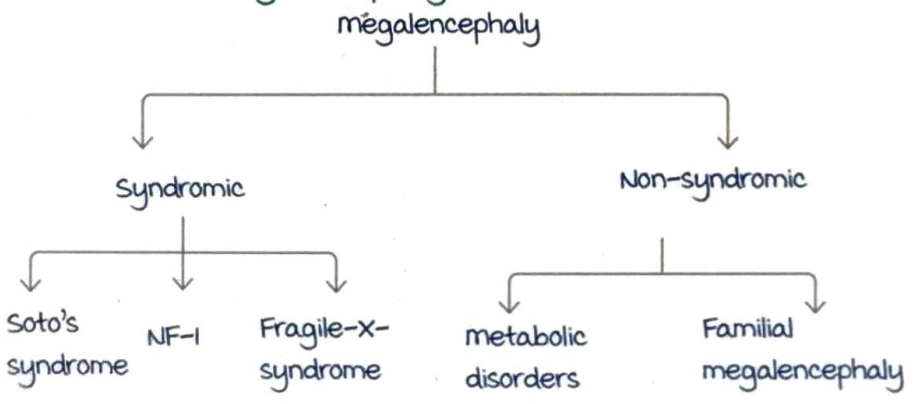
Causes of macrocephaly :

- Increased amount of CSF :
    1. Hydrocephalus.
    2. Hydranencephaly : Absent brain tissue is replaced by sacs containing fluid (CSF). Transillumination (illiminate upwards from below the skull) is positive.
- Increased bleeding within skull :
    1. Sub dural hemorrhage.
    2. Extra dural hemorrhage.
    3. Intraventricular hemorrhage.
    4. Sub arachnoid hemorrhage.
- Increase in size of bony compartment :

Associated with primary bone disorders like :

1. Achondroplasia.
2. Osteogenesis imperfecta.
3. Abnormal skull expansion : Beta thalassemia major (skull also involved in erythropoiesis).

- Increased brain tissue :
    1. Megalencephaly



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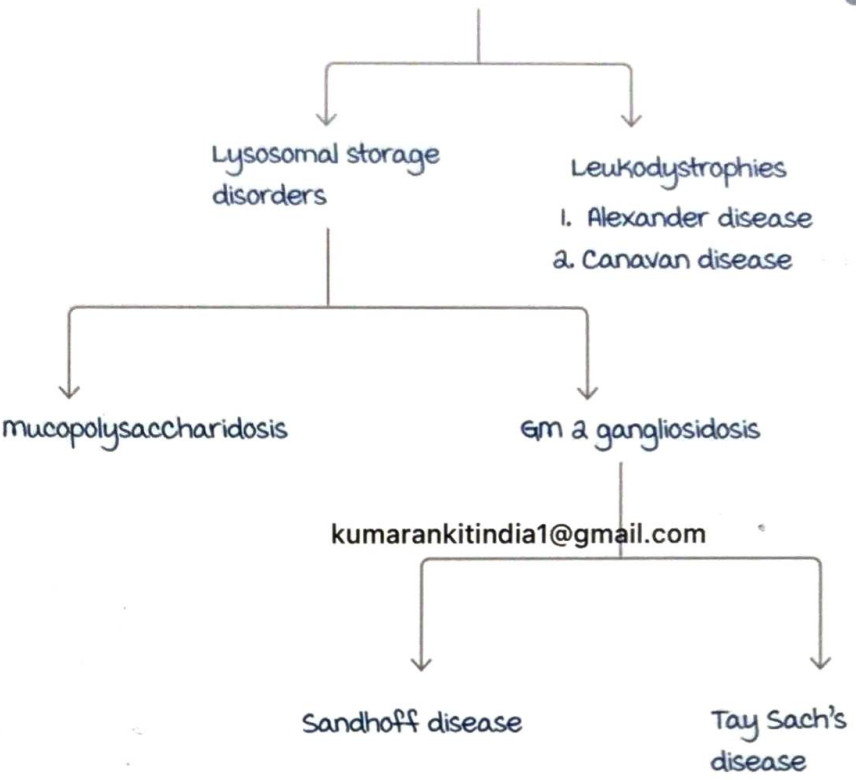

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Soto's syndrome (cerebral gigantism) : Increase in head size and height are noted.

Fragile X Syndrome (CGG trinucleotide repeat) :

- Low IQ.
- Macro-Orchidism.
- Elongated face and large ears.

NF-1 (Neurofibromatosis -1) : Neurocutaneous syndrome.

Familial megalencephaly :
Normal IQ and normal development seen. All family members have large heads.

metabolic disorders : Patients have low IQ and delayed development.

Tay Sach's disease : Hexosaminidase A deficiency.
No hepatosplenomegaly is seen.
Sandhoff disease : Hexosaminidase A and B deficiency.
Hepatosplenomegaly is seen.

Both Tay Sach's and Sandhoff disease have cherry red spots in the macula.

Active space

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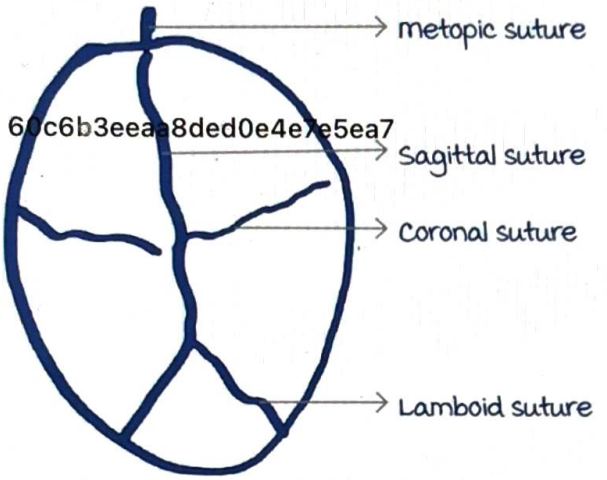
Craniosynostosis

00:37:12

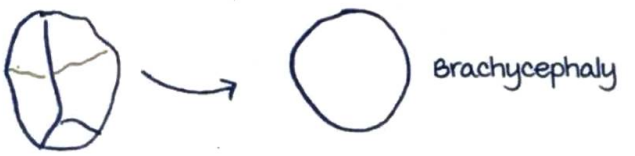
Synostosis : fusion, Cranio : skull.
In this condition, there is premature fusion of sutures.
Growing brain needs space → skull expands in an irregular manner.

Whenever the suture fuses, the skull elongates along the direction of the fused suture.
most common suture to undergo premature fusion : Sagittal suture.




Posterior plagiocephaly


Anterior plagiocephaly


Brachycephaly


Dolichocephaly/
Scaphocephaly

Active space

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Premature fusion of	Craniosynostosis
Metopic suture	Trigonocephaly (in front of skull).
Unilateral coronal suture	Anterior plagiocephaly
Bilateral coronal suture	Brachycephaly
unilateral lambdoid suture	Posterior plagiocephaly
Sagittal suture	Dolicho/scaphocephaly (↑ AP diameter)

Brachycephaly is a characteristic type of skull noted in down syndrome (broad skull with increased Bi-parietal diameter).

Syndromic associations with craniosynostosis :

- Crouzon syndrome :
    1. Autosomal dominant condition.
    2. Proptosis (underdeveloped and shallow orbital bone). Prominent feature.
    3. Hypertelorism (underdeveloped orbit).
    4. Maxillary hypoplasia.
    5. Bilateral coronal suture fusion : Brachycephaly.
    6. Normal IQ.

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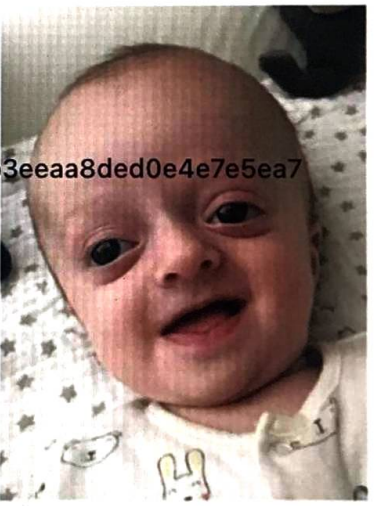

Crouzon Syndrome

- Apert Syndrome :
    1. Autosomal dominant condition.
    2. Not so prominent proptosis.
    3. Asymmetric face.
    4. Syndactyly or mitten hand deformity.
    5. Cleft lip/palate.
    6. Low IQ.

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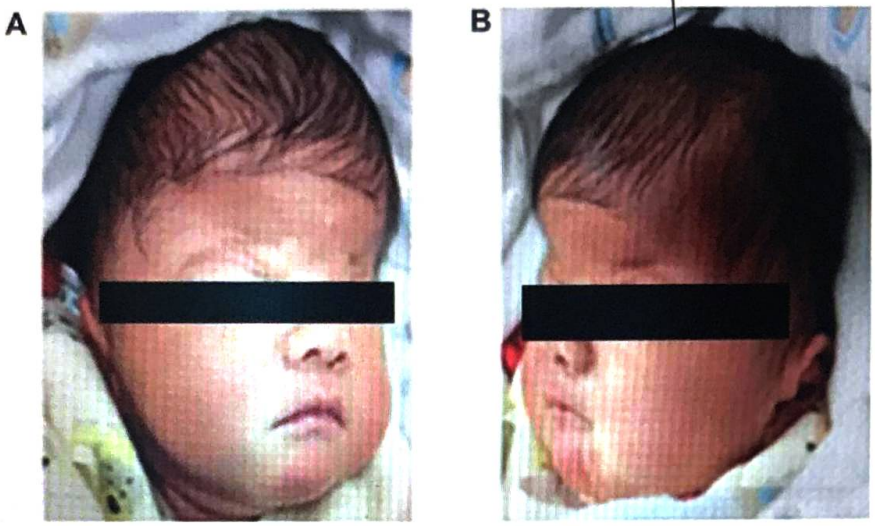
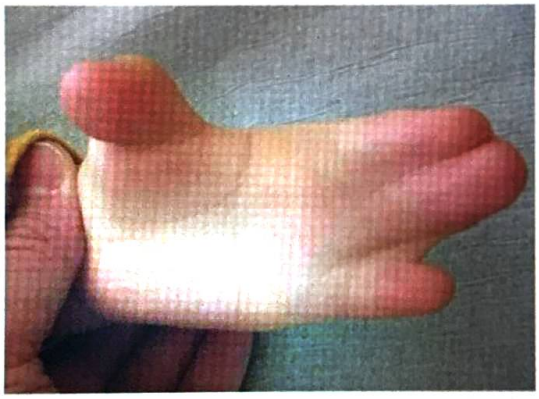
7. Multiple sutures prematurely fuse :

Turricephaly or oxycephaly or acrocephaly.

(Tower shaped skull/conical skull)



Apert Syndrome



Syndactyly/ mitten hand deformity.

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SHORT STATURE IN CHILDREN

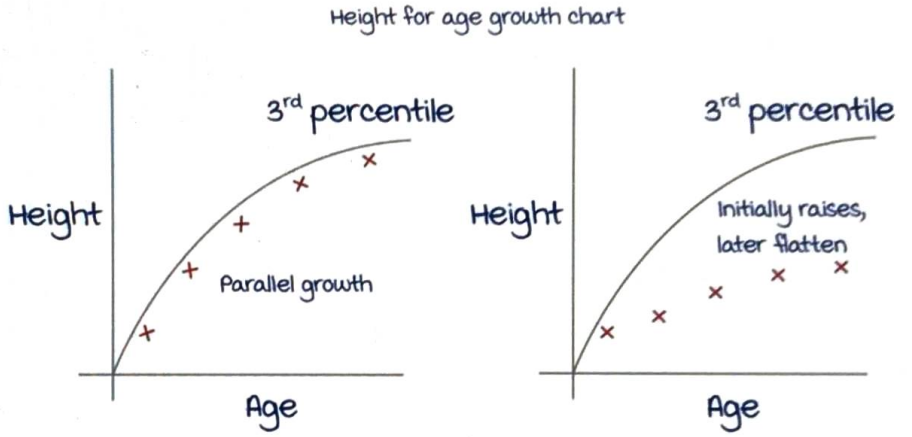
Introduction

00:00:36

It is defined as height for age < 3rd percentile or < (-2 SD).

Types

Normal variant short stature >> Pathological short stature



Growth velocity :

It is the rate of increase in height.

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It is normal in normal variant of short stature but in pathological variant, it is decreased or absent.

If growth velocity is < 25th percentile (very low growth velocity) then it is a pathological variant but if it is > 25th percentile, then its normal variant short stature.

Normal variant short stature

00:04:36

Types

Constitutional delay >> Familial short stature

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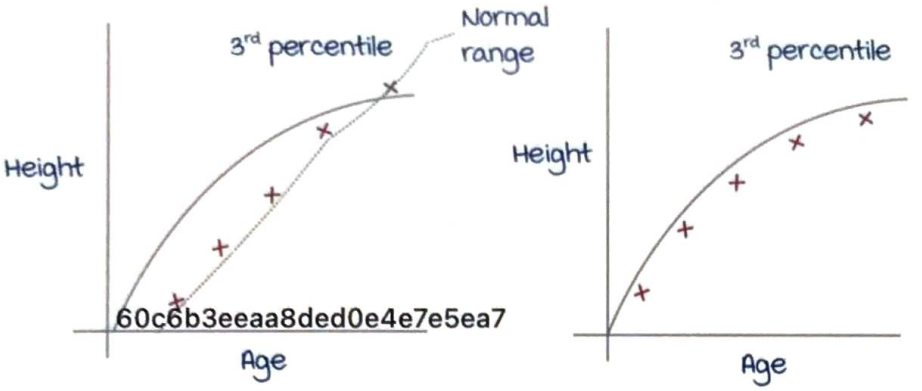

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X = Value of height in a child at different ages.

Features	Constitutional delay	Familial short stature
Expected adult height	Normal	Short
Parent's height	Normal + h/o short stature in childhood	Short
Puberty	Delayed	At normal age
Bone age	Delayed	Normal

Constitutional delay is the overall MCC of short stature & delayed puberty in children : Constitutional delay aka constitutional delay in growth & puberty.

Pathological short stature

00:08:23

Causes :

- Chronic malnutrition : Stunting.
- Bone disorders :
  Achondroplasia, osteogenesis imperfecta, rickets.
- Genetic syndromes :
  Trisomies (E.g. Down's syndrome), turner syndrome.
- Hormonal disturbances :
  Deficiencies of thyroid hormone, growth hormone, parathyroid hormone in hypoparathyroidism, delayed puberty by deficiency of gonadal steroids like estrogen & testosterone.
  Excess hormone : Cushing syndrome (excess

Active space

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glucocorticoids. short stature - prominent manifestation).
Precocious puberty (early puberty → excess gondal steroids → premature fusion of epiphysis → growth arrested at an age earlier than expected).

Approach to short stature

00:11:44

1. Using body proprotions :
In relation to upper segment (US) & lower body segment (LS).
Helps in differentiating short stature into proportionate and disproportionate short stature .

Proportionate short stature :

US : LS is normal for age.

Causes are :

- All normal variants (constitutional & familial variants)
- Malnutrition.
- Chronic systemic illness (CKD, CLD, cardiac problem)
- Growth hormone deficiency.

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Disproportionate short stature :

- US : LS is abnormal for age.

Types :

1. Short trunk/Short US : Small TV.
   - Spondyloepiphyseal dysplasia.
   - Mucopolysaccharidoses.
   - TB (Pott's) spine.
   - Vertebral anomalies (e.g. hemivertebrae, butterf shaped vertebra).

2. Short limb/Short LS :
   - Achondroplasia.
   - Rickets.
   - Congenital hypothyroidism.

2. Bone age determination :
Using ossification centres in the bone, seen on X-ray.

Active space

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- 1st year : Shoulder.
- 1 to 13 years : Left Wrist (look at carpal bones).
- 12 to 14 years : Elbow and hip.

Methods for bone age estimation :

- Tanner Whitehouse method.
- Gruelich Pyle Atlas (X-rays for different age groups of males & females are available.
  X-ray of patient is matched with X-ray on the atlas & age is determined).

Delay in bone age	Advanced bone age
1. Constitutional delay 2. Malnutrition 3. Hormone deficiency as hormones are needed for ossification centre appearance.	Precocious puberty (excess of gonadal steroids).

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Q. You are evaluating a child with pathological short stature, abnormal facies and short trunk. Physical appearance reveals the following finding. Which of the following disorders should be suspected ?

A. Achondroplasia.
B. MPS.
C. Hypothyroidism.
D. Rickets.

Active space

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Mucopolysaccharidoses

00:22:07

Lysosomal storage disorder.
Defect in lysosomal enzymes (hydrolases) causes accumulation of Glycosaminoglycans (GAG) Eg : Heparan sulphate, chondroitin sulphate, dermatan sulphate.

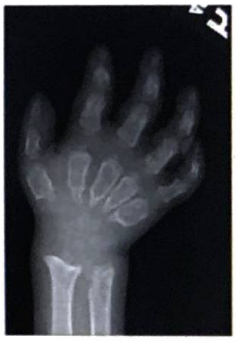
Characteristic abnormal facies called Gargoylic facies.

- Broad forehead.
- Upturned nose with copious nasal discharge.
- Macroglossia.

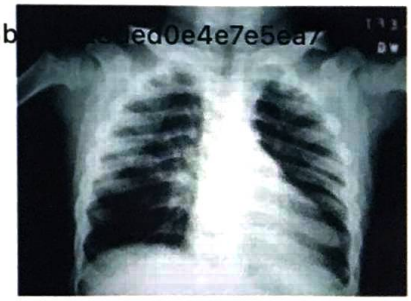
Cloudy cornea (whitish cornea).
Low IQ, Macrocephaly.
Abdominal distension with increased risk of umbilical and inguinal hernia.
Hepatosplenomegaly.

Radiological findings :

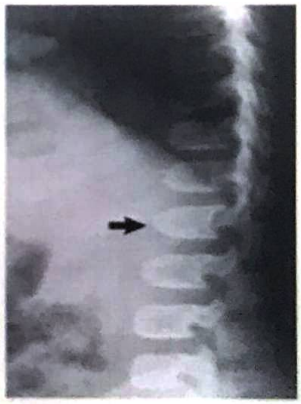
- Bullet shaped metacarpals (proximal narrowing).



- Spatula/oar/paddle shaped anterior ribs : Flat straigh ribs.



Narrowing/beaking of the vertebra.



Active space

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Short trunk is seen because of narrowing of multiple vertebrae.

MPS is associated with multiple bony abnormalities together called Dysostosis multiplex.

Type	Name	Enzyme deficiency
MPS I MPS IS	Hurler syndrome Scheie syndrome (Formerly MPS type V)	L-Iduronidase.
MPS II	Hunter syndrome	Iduronate sulfatase
MPS III	Sanfilippo (mc)	Heparan sulfamidase
MPS IV	Morquio	N- acetyl galactosamine sulfatase
MPS VI	Maroteaux Lamy	Aryl sulfatase B
MPS VII	Sly	β- glucuronidase

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Hurler syndrome : Complete deficiency of L-Iduronidase.
Scheie disease : Partial deficiency of L-Iduronidase.

All MPS are inherited Autosomal recessively except,
Hunter syndrome : X linked recessive.
Hunter syndrome : No corneal clouding.
Morquio syndrome : Highest chance of bony deformities.

Low incidence of intellectual disability : IS, IV, VI.
Enzyme Replacement Therapy (ERT) can be given for MPS I, II, IV, VI.

Achondroplasia 00:33:34

Characterized by short limbs & short stature.

- Proximal limb shortening (arm > forearm) called rhizomelia.
- Mostly inherited as autosomal dominant.
- FGFR 3 (Fibroblast Growth Factor Receptor) gene mutation (chromosome 4p).

Active space

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Other features :

- Macrocephaly (large head) because of small foramen magnum causing obstructed flow of CSF leading to hydrocephalus.
- Broad & bulging forehead.

X-ray pelvis shows champagne glass appearance.

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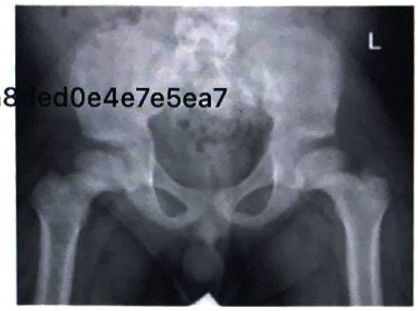
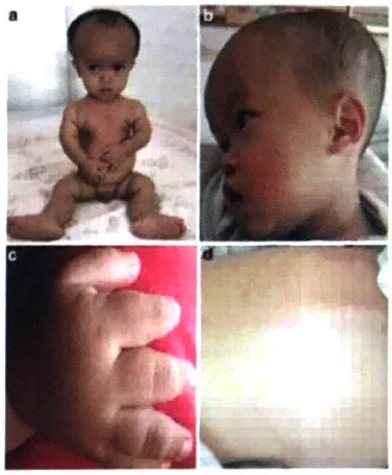


Champagne glass pelvis



Achondroplasia

Active space

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NORMAL DEVELOPMENT IN CHILDREN

Developmental milestones :

Defined according to 4 areas or domains of development.

- Gross motor (major motor activities).
- Fine motor (usage of small muscles of hands).
- Language (communication with others).
- Social (other modes of interactions).

Gross motor milestones

00:02:56

Rule : Development in a child always proceeds in cephalocaudal direction (head - foot).

3 months : Head control/neck holding (1st milestone). 60c6b3eeaa8ded0e4e7e5ea7

4-6 months : Roll over (supine to prone). Trunk control present.

6 months : Sit with support (tripod position : leaning forward trunk in centre with two arms by the side).

Tripod position

8 months : Sit without support, crawling.

Crawling

Body is flat on the ground and uses both upper limbs and lower limbs to move from one place to another.

Active space

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10 months : Standing with support, creeping.

Creeping

Creeping : Uses all 4 limbs to move but chest and abdomen are lifted above the ground.

11 months : Cruising.

Cruising with support

Cruising : Uses support to move sideways.

12 months : Standing without support, walking with support.
15 months : Walks without support/independently.
18 months : Running.
2 years : Climbing stairs with 2 feet/step holding side rails.
3 years : Climbing stairs with 1 foot/step (upstairs), rides tricycle.
4 years : Upstairs & downstairs – 1 foot/step, hopping.

Hopping (jumping on one leg)

Active space

Leave Feedbac

Fine motor milestones

00:12:38

Small movements of hands like grasping objects, drawing skills, dressing skills and making tower of cubes.

0–3 months : No fine motor milestone (babies hands are always closed due to neonatal palmar grasp reflex).

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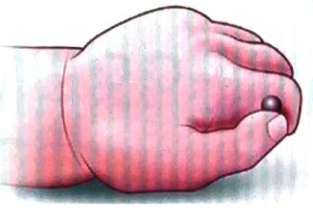
4 months : Reaching out for objects with both hands (bidextrous grasp).

6 months : Unidextrous grasping (ulnar palmar grasping – immature) Child uses palm as support ; transfer objects from one hand to another.

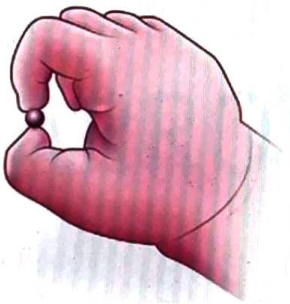
8 months : Radial palmar grasping : mature.

9 months : Immature pincer grasp : Object held by sides of fingers.



Immature pincer grasp (9 months)

12 months : Mature pincer grasp : Object held by finger tips.



Mature pincer grasp (12 months)

15 months : Scribble.

18 months–2years : Draw a vertical line.

2 – 2 1/2 years : Draw a horizontal line.

Best answer for drawing a line : 2 years (if horizontal or vertical line is not mentioned).

Active space

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2 years : Line (L).
3 years : Circle (O).
4 years : Cross/plus (X).
4 1/a years : Square (S).
5 years : Triangle (T).
6-7 years : Diamond/rhomboid (D).

Mnemonic : LOX STD

Undressing : 2 years (easier task than dressing).
Dressing : 3 years.
Both these requires help of a parent.
Dresses and undresses without any help at 5 years.

Tower of cubes : Placing one cube on top of another.
Starts at 15 months : 2 cubes.
18 months : 3 cubes.
24 months (2 years) : 6 cubes.
36 months (3 years) : 9 cubes.

Language milstones

00:24:36

Comprises of tasks like making sounds, talking words/sentences, telling stories/rhymes etc.

Starts from 3 months.
3 months : Musical sounds when child is happy : Cooing sounds.
4 months : Laughs aloud when happy.
6 months : Monosyllables (ma, pa).
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9 months : Bisyllables/repeating same syllable (mama, papa).
1 year : 1-2 words with meaning.
15 months : Jargon speech (words without meaning is temporary milestone for 1-2 months).
18 months : 8-10 words with meaning.
2 years : 100 words ; starts to speak in sentences.
3 years : Recognize and tell name, age and gender.
4 years : Story telling or singing rhymes.

Active space

Leave Feedbac

Social milestones 00:30:00

2 months : Social smile.
3 months : Mother regard (interacts with mother only).
6 months : Stranger anxiety (cries) ; smiles at mirror image.
9 months : Waves bye-bye.
12 months : Playing a simple ball game with others.
15 months : Points to object (asking for a ball far away).
18 months : Domestic mimicry (child imitates actions of adults).
2 $_{1/2}$ – 3 years : (Non-interactive) parallel play.
4 years : (Interactive) group play.
5 years :

- Follow 3 step commands (go to room, take the ball, keep it here).
- Identify 4 colours and repeat 4 digits.

Miscellaneous milestones 00:35:36

- Mouthing : Starts 6 months ; Put objects/hand in mouth.
- Casting : Begins by 1 year of age (deliberate throwing of objects).
  Both milestones are seen till 18 months–2 years.

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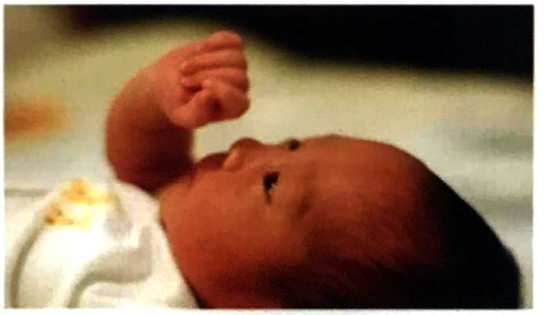
- Object permanence : Around 9 months.
  Feeling that a missing object is present around the child though not seen in the field of vision.
  E.g : Looking and searching for a ball that fell off from a child's hand.
- Handedness : Preference of one hand over the other.
  Starts by 3 years & firmly established at 4 years.
- Hand regard : Appears at 3 months and persists till 5 months.



Active space

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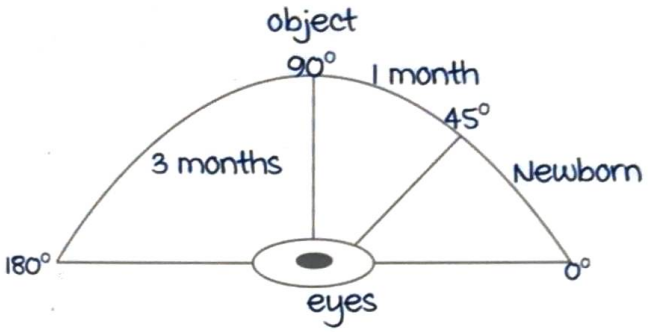
Abnormally persisting hand regard for more than 5 months : Developmental delay.

- Range of vision :

Newborn follow objects upto 45°.
1 month child follow objects upto 90°.
3 months old child follow objects upto 180°.
Binocular vision fully established by 4 months.



- Localization of sound : Turning head towards sound source.
- Assesed using Murphy's sequence.

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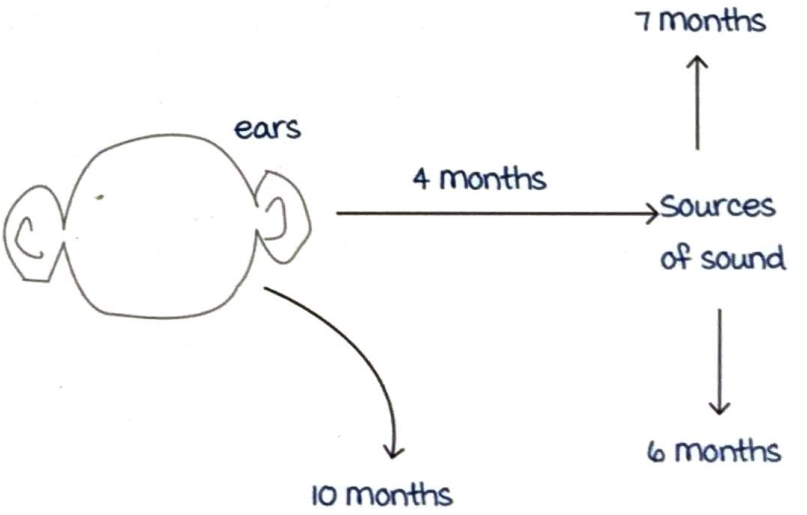
Newborn : Startle response to sound.
4 months : Horizontal localization of sound.
6 months : Downward localization of sound.
7 months : Upward localization of sound.
10 months : Diagonal localization of sound.



Active space

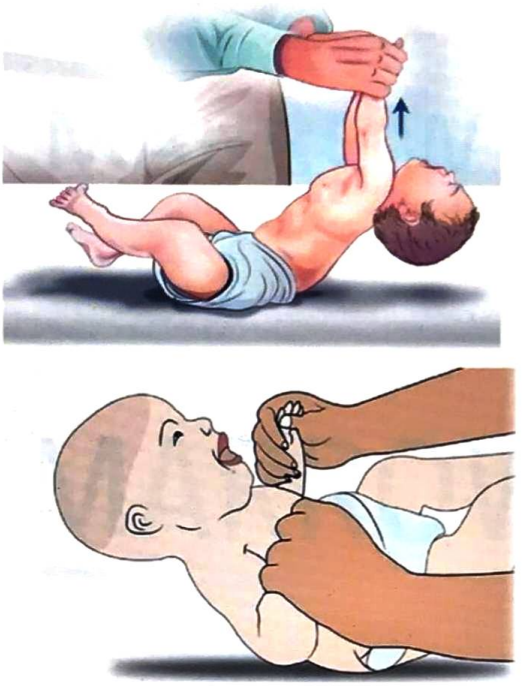
Assessment in development

00:46:50

Pull to sit maneuver :

Newborn : Complete head lag (no head control).

3 months : No head lag (head control attained).



Normal pull to sit

Ventral suspension :

Lift the child using palm of a hand & turn to prone position with palm still supporting the baby's trunk.

Newborn : Head and limbs drop down (no control).

2 months : Head at level with rest of body.

3 months : Head is lifted above the level of body.

Prone position :

Newborn : Same posture till 2 weeks of life.

- Legs folded and kept beneath abdomen.
- Pelvis : Lifted above the rest of body.

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Active space

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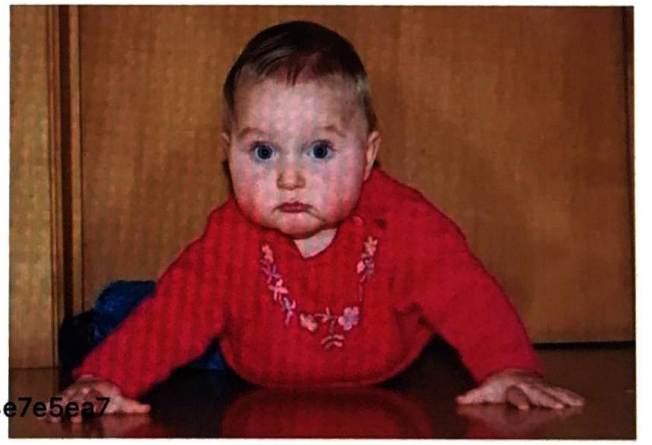
4–6 weeks : Legs straighten and pelvis at level with rest of body (baby will lie flat).

3 months :

- Head lifted up.
- Elbow flexed/bent.
- Bears weight on forearm.

6 months :

- Head lifted up.
- Elbow straight (in extension).
- Bearing weight on extended limbs.



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DISORDERS OF DEVELOPMENT

Assessment of development of the child 00:00:11

$$\text{Developmental quotient (DQ)} = \frac{\text{Developmental age}}{\text{Chronological age}} \times 100$$

Eg : A 6 year old child is able to draw only a circle. Therefore the developmental age of the child is 3 years. DQ = 50.

DQ < 70 : Abnormal / Developmental delay

DQ < 70 in ≥ 2 domains : Global developmental delay

Evaluation of a preterm baby :

- Born at 32 weeks of gestation (premature by 2 months, as normal term is at 40 weeks). Preterm correction = 2 months.
- Evaluated at 3 months of age (Postnatal age)
- Corrected age = Postnatal age − Preterm correction. Here 1 month/ the child is developmentally considered as 1 month old.
- Correction for prematurity can be calculated till 2 years of age.

Red flag signs of development 00:05:59

When any of the following milestones crosses the upper limit to develop, it is considered a red flag sign.

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Milestone	Upper limit
Visual fixation or following	2 months
Head control	5 months
Vocalization	6 months
Sitting without support	10 months
Standing with support	12 months
Standing/walking without support Single words	18 months

Active space

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Tests for developmental assessment 00:07:56

Screening tests : Mnemonic - Good Doctor Treats Patients

1. Goodenough-Harris 'draw a man' test
2. Denver II
3. Trivandrum development screening test
4. Phatak's Baroda screening test

Definitive tests :

1. Bayley II scale (Infant and toddler development)
2. Stanford Binet Intelligence scale
3. Weschler Intelligence scale
4. Vineland adaptive behaviour scale II

Developmental delay in a child 00:10:03

The causes for developmental delay can be classified as :

Congenital	Acquired
Chromosomal (eg., Down's)	Severe head trauma
Genetic Syndrome Eg : Fragile X syndrome Red syndrome	Meningitis
Congenital hypothyroidism	Perinatal asphyxia
Brain anomalies	
Torch infections	

Variants of developmental disorders :

- Developmental dissociation :
  Variation in attainment of milestones between 2 or more domains.
  Eg : Isolated language delay due to hearing deficit.
- Developmental deviance :
  Child develops milestones out of normal sequence.
  Eg : Early rolling over due to increase tone of extensor muscle in cerebral palsy.

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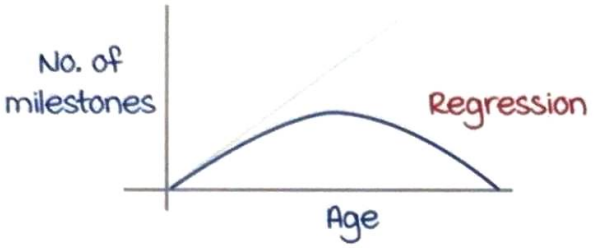
Regression :

The child loses previously acquired milestone.



Eg : Rett syndrome

Subacute sclerosing panencephalitis (SSPE)

Leukodystrophy

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BEHAVIOURAL DISORDERS IN CHILDREN

Breath holding spells 00:00:40

It is a reflex event.

Age : Starts after 6 months.

Peaks by 2 years of age and persists till 5 years.

Sequence of events :

Provocative factors like anger, frustration or pain

↓

Cry

↓

Starts to hold breath

Breath holding takes in full expiration.

Two types :

1. Most common : Due to increase in parasympathetic activity.
   Cyanosis is developed.
   Child becomes normal after few seconds.
   May develop a tonic clonic movement.
2. Increase in sympathetic activity (Palid type) :
   Child becomes pale and white mimicking a syncope like attack.
   It usually lasts for few seconds.
   If it lasts longer, ECG is done to rule out long QT syndrome.

Management :

- Reassurance and inform parents about benign nature and spontaneous resolution.
- During tonic clonic episode : Child should be turned to one side (prevents aspiration).

Active space

- Never pick up a child during the event (can abruptly decrease cerebral perfusion).

Should do work up for Iron deficiency anaemia and treated accordingly.

Atropine : Useful in cases of long duration palid spells.

Bruxism :

Teeth grinding.

It is common up to 30% of children.

Age : Less than 5 years (normal).

Commonly occurring during sleep (normal phenomenon).

Daytime bruxism (>5 years) : associated with anxiety disorders.

Long term problems : Significant dental malocclusion, jaw pain (Temporomandibular joint pain).

Older children (>5 years) : may benefit from behavioural therapy.

Pica

00:09:52

Consumption of inedible/non nutritive substances like chalk, mud or paint by a child for a period of atleast 1 month.

It should also be inappropriate for development or cultural practices.

Age : < 5 years.

Risk factors : Malnutrition, iron deficiency anemia, low socioeconomic status, psychosocial stress (parenteral neglect, maternal deprivation) and developmental delay (Cerebral palsy).

Complications :

- Lead poisoning (paint).
- Parasitic infestations (mud) 60c6b3eeaa8ded0e4e7e5ea7

Management : Behavioural therapy.

One dose of deworming (Albendazole).

Low dose iron supplements (0.5 to 1 mg/kg/day)

Active space

Leave Feedba

Thumb sucking

00:15:20

Self soothing behaviour for a child.
Noted in the age group after 6 months.
Peak : 18 to 21 months.
Till 4 years, it is normal (only reassurance is required).
>4 years : Emotional stress (also a sign of social insecurity).

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Management :
>4 years : Behavioural modifications (positive reinforcement by praising the child when he is not sucking the thumb).
Negative reinforcement (like applying oil) is not recommended.

Temper tantrums :
Starts in the age group of 18 to 36 months.
Persists till 6 years.
It is response of child to physical or emotional challenges

↓

Crying/ kicking/ pushing/ head banging.

It is a attention seeking tactic of the child.

Management :
Reassure and parents and ask them to remain calm during the time of episode.
The child should be left alone in a place where he or she is unlikely to get harmed.
Distraction (Taking away the child from that place)/time out technique (leaving the child alone for sometime to settle down) can also be implemented.

Active space

Tics

00:21:02

It is considered as fast involuntary paroxysmal non rhythmic repetitive motor or verbal manifestations.
Occurs as abrupt onset.

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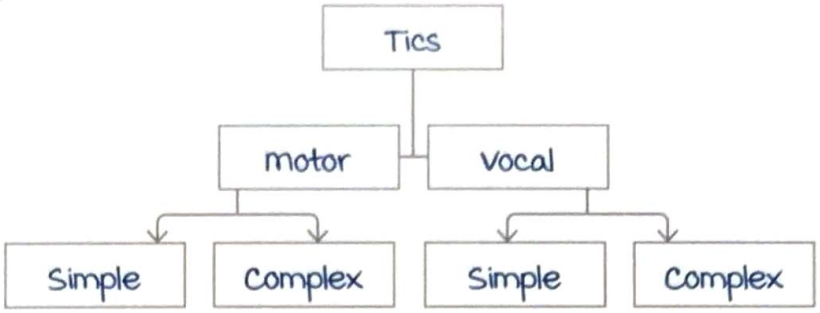
Types :



Simple motor tics : Eye blinking, neck jerking and shoulder shrugging.
Complex motor tics : Echopraxia (repeating movements or postures/ imitating others).

Simple vocal tics : Throat clearing, cough, sniffing.
Complex vocal tics : 1. Echolalia (repeating other's speech),
2. Paralalia (repeating one's own words/ sentences).

Age groups : Starts by 4 to 6 years.
Peak : 10 to 12 years.
Persists till 18 to 20 years.

Tourette's syndrome :

Age of onset < 18 years.
Both motor and vocal tics for >/= 1 year.

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management :

Behavioural therapy.
Long lasting tics : Neuroleptics (Haloperidol, Clonidine).

Nocturnal enuresis 00:27:02

Urination at night time after the age of > 5 years (age of bladder maturation).
It is nearly complete evacuation of bladder at a wrong place & time for at least twice a month for at least 3 months.
Males > Females.
Types :

- Primary (m/c) : Present since birth.

Active space

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- It is a functional disorder.
- Delayed bladder maturation.
- Few cases : Decreased ADH secretion at night.

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- Secondary : Child was previously dry at night for at least a period of preceding 6 months.
  Causes : UTI, Diabetes insipidus and mellitus, stress, bowel bladder dysfunction.

Other types : Based on symptomatology.

- Monosymptomatic (m/c) : Only nocturnal enuresis.
- Polysymptomatic :
  Nocturnal enuresis with Lower Urinary Tract Symptoms (LUTS) like hesitancy, urgency & dribbling of urine.

Most common variety of nocturnal enuresis : Primary monosymptomatic nocturnal enuresis.

Management :
Primary monosymptomatic nocturnal enuresis : Reassurance.
Behavioural modifications : Voiding before going to bed, restrict fluids in the evening (40% fluids in morning, 40% at noon and 20% after 6 pm in the evening).
Avoid caffeinated drinks (tea, coffee and soda).

Motivational therapy : Verbal praising, gifts for being dry at night.
Alarm therapy : Form of conditioning a child to sensation of full bladder (attaching sensors to underwear or sleeping mat of the child).
Simple alarms at fixed timings can also help.
Motivational therapy + Alarm therapy : Best with 60 to 70% success rate.
If all the above therapies fails : Drug therapy
Short term : Oral desmopressin.
Long term usage : Oxybutynin/Tolterodine (decrease uninhibited bladder contractions).

Imipramine are not used because of adverse cardiovascular side effects.

Active space

107
Leave Feedba

BREAST FEEDING

Breastfeeding basics 00:00:38

Breastfeeding should be started as soon as possible.
Exclusive breastfeeding should be continued till 6 months and Top feeding should not be practised.
Breast milk contains 88% of water.
After 6 months, complementary feeds (semisolids→ solid) are started.
Breastfeeding should be continued till 2 years of age along with complementary feeds.
Maternal reflexes :
Prolactin reflex :

- Prolactin helps in the production of breast milk : galactopoiesis.
- Active at night time (mother should not skip night feeds to maintain prolactin reflex).

Oxytocin reflex :

- Oxytocin acts on myoepithelial cells causing contraction. Helps in ejection of milk : galactokinesis.
- Active all the time.
- Affected by maternal emotions.

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Neonatal reflexes :
Rooting reflex :
When a newborn baby's mouth touches the nipple at the start of breastfeeding, newborn turns the mouth towards the stimulus. Helps in initiation of feeding.

Suckling and swallowing reflex :
When the nipple region touches the baby's palate, baby starts the process of sucking. Once the baby's mouth gets filled with milk, swallowing reflex is activated, and baby swallows the milk.

Suck – swallow – breathe cycle during breastfeeding is for approximately 1 second.
Correct infant latch – on position :

Active space

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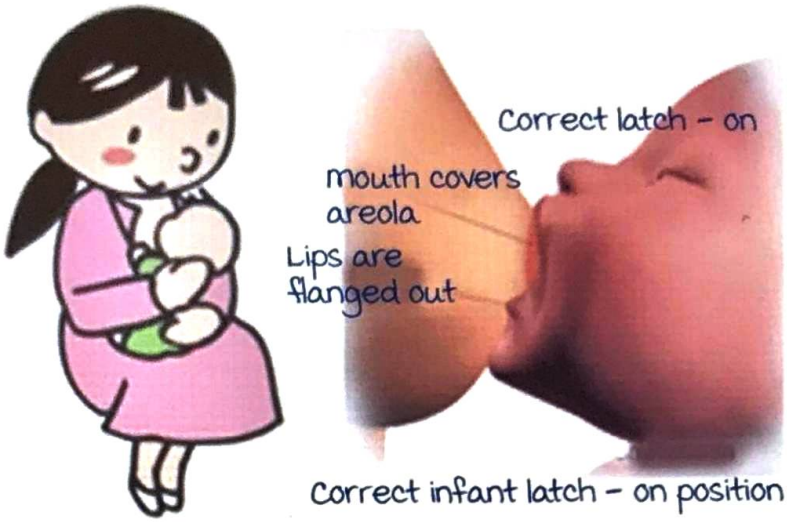

Correct infant latch - on position

Features of good attachment (latching) :

- Baby's mouth is wide open.
- Areola and nipple in baby's mouth. Upper part of areola may be visible but lower part should be completely covered by baby's mouth.
- Lips are everted.
- Chin of the baby touches the mother's breast.

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Properties of breast milk

00:10:08

Immunological advantages :

Predominant immunoglobulin : Ig A (secretory Ig A).

Others : (mnemonic PLAB).

P : Low levels of Para amino benzoic acid(PABA) → protects against malaria as the parasite requires it for replication.

L : Lactoferrin → protects from E.coli infection.

A : Ig A

B : Bifidus factor → protects from E.coli infection.

Bile salt stimulated lipase → protects against giardiasis.

Bioactive factors :

Epidermal growth factor (EGF) & Transforming Growth Factor β (TGF β).

They enhance the integrity or maturity of gastrointestinal epithelium.

Decrease the risk of allergy in later life.

Active space

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Nutritional properties :

Nutritive value : 67 Kcal/100 ml (same for cow's milk).

Carbohydrates :

6-7 g/dl → high compared to cow/buffalo's milk.

Majority of carbohydrate is lactose.

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Lactose :

- Lactase breaks lactose into glucose and galactose.
- Galactose is further broken down into glucose.
- Lactose helps in the production of lactoferrin →protects against E.coli infection.
- Lactose helps in the colonization of GI tract by good bacteria like lactobacillus.

Galactocerebroside (Galacto oligosaccharide) :

Important for nutrition of oligodendrocytes (helps in myelination, brain growth).

Fat :

3.5 - 4 g/dl → Almost same as in cow milk.

Rich in polyunsaturated fatty acids (PUFA).

Eg : Arachidonic acid (ARA), docosahexaenoic acid (DHA).

Advantages : Favour brain growth.

Protein :

0.9 - 1.1 g/dl → 3 times less compared to cow's milk.

Quality of protein is superior.

Breast milk	Cow's milk
Whey predominant.	Casein predominant.
Ratio of whey : casein is 60 : 40.	Ratio of whey : casein is 20 : 80.
Whey protein is easily digestible.	Casein is poorly digestible → undergoes coagulation in the intestine → Constipation.

Examples of whey protein in breast milk : Lactalbumin, lactoglobulin. They favour brain growth.

Active space

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Amino acids promoting brain growth : Taurine, cysteine.
Increased risk of allergy with cow milk protein → Cow's milk protein allergy (CMPA).

Other benefits of breastfeeding :

- Protection from illness /infection : 14 times less likely to die of diarrhea and 4 times less likely to die of respiratory infection compared to a non breast fed baby.
- Allergy : Decreased risk of allergy.
- Decreased risk of diabetes & heart disease in later life.
- IQ : Baby who is breastfed till the age of 2 years has a higher IQ compared to baby who is not breastfed till the age of 2 years.

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Benefits for mother :

- Helps in uterine involution.
- Decreased incidence of postpartum hemorrhage.
- Lactational amenorrhea for around 6 months.
- Decreased risk of carcinoma breast & ovary.
- Shedding weight after pregnancy.

Deficiencies in breast milk

00:25:48

- Micronutrients deficient :
  Vitamin K,
  Vitamin D,
  Vitamin B12 (found in pure vegan/vegetarian families).
- Others : Iron. Though iron levels are low, iron has good bioavailability and baby has enough iron stores from mother. So, routine iron supplementation is not required.

Composition of breast milk :

- Colostrum : Milk secreted in the first 3 – 4 days. Thick, yellowish with high fat content & immunoglobulins (IgA).
- Transitional milk : Milk secreted till 14 days after birth. Protein content and immunoglobulins decrease, fat and sugar content increase.
- Mature milk : After 14 days.

Active space

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Parts of breast milk : Foremilk (whitish) and hindmilk (yellowish).

Foremilk is rich in water. It satisfies the thirst in the child.
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Hindmilk is rich in fat. It satisfies the satiety (hunger demand) of the child.

Preterm milk :
Milk secreted by mother who has delivered a preterm baby.
Contains more sodium, iron, protein and calories (sugars).
(Mnemonic : Preterm baby drinks in SIPS.)

Contraindications of breastfeeding :
Absolute :

- Lactose intolerance (congenital Lactase deficiency).
- Galactosemia.
- Mother on chemo/radiotherapy.

Relative :
Infections in mother :

- HIV (mother has to take antiretroviral treatment and baby has to take Nevirapine prophylaxis). Ideal is formula milk if affordable.
- Tuberculosis if the mother is not on antitubercular treatment or has competed <2 weeks.
- Herpes simplex and herpes zoster if the mother has lesions on the breast.

COVID infection in mother is not a contraindication for breastfeeding.

Expressed breast milk storage :

- Room temperature : 6 – 8 hours.
- Refrigerator : 24 hours.
- Freezer (– 20° C) : 3 months.

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MALNUTRITION

Classification of malnutrition 00:00:28

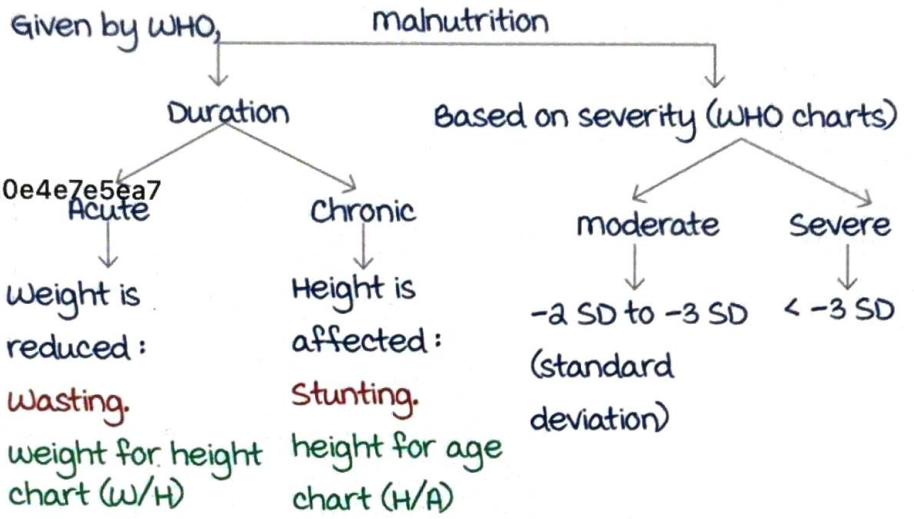

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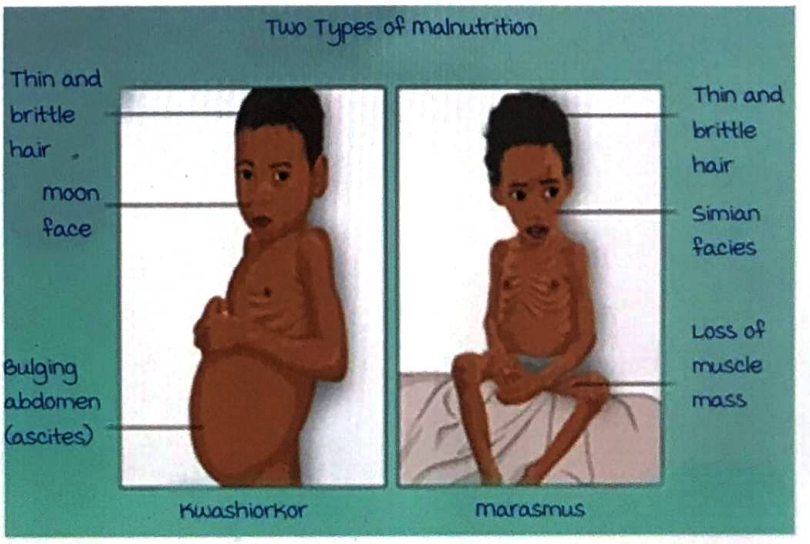
chart chart

	moderate malnutrition	Severe malnutrition
Symmetrical edema (B/L pedal edema)	No	Yes
Weight for height (acute)	-2 to -3 SD	< -3 SD
Height for age (chronic)	-2 to -3 SD	< -3 SD

Extreme forms of malnutrition (PEM/PCM) 00:05:08



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Kwashiorkor :

Main sign is pitting edema.

Predominant protein deficiency → albumin reduced → decrease in oncotic pressure → edema.

Mediated by free radical damage.

Decompensated malnutrition/ dysadaptative malnutrition.

Onset : 1 – 4 years.

General appearance :

- Dull/ lethargic appearance.
- Poor appetite.

Muscle wasting is present, masked by edema. 60c6b3eeaa8ded0e4e7e5ea7

Also called fat sugar baby.

Skin changes :

- Areas of hyperpigmentation and areas of dyspigmentation called as Flaky paint dermatosis (confluent areas) and enamel spots (isolated spots).
- Crazy pavement dermatosis : Cracks on skin surface.

Hair changes :

- Patchy pigmentation, few areas of pigmentation with areas of no pigmentation, called flag sign.
- Brittle, lustreless, easily pluckable hair, loss of curls. Fatty liver is present.

Marasmus 00:14:06

More common compared to Kwashiorkor.

Caused by severe deficiency of calories (predominant).

Prominent and visible wasting (main sign).

Wasting is due to adaptation to starvation, hence called adaptive starvation (mobilization of fat, results in fat loss).

Skin & bone appearance is seen.

Simian facies (monkey like face).

Baggy pant appearance (lot of skin folds, due to loss of fat and muscle).

General appearance : Alert, good appetite.

Outcome is better as they have good appetite.

Active space

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Onset : < 1year.

	Kwashiorkor	Marasmus
Predominant deficiency	Protein	Calories
Onset	1 – 4 years	< 1 year
Edema	+++	Absent
Muscle wasting	+ (hidden)	+ visible severe
Appetite	Poor	Good
General appearance	Lethargic, apathy	Alert
Additional features	Hair changes (flag sign). Skin changes. Presence of fatty liver.	

Severe acute malnutrition (SAM) 00:20:13

Definition :

1. WFH < -3 SD
2. Bipedal edema (diagnosis of exclusion)
3. Mid arm circumference ≤ 11.5 cm (age 6 months – 5 years).

Presence of any 1 of these criteria is enough to term as SAM.

	Uncomplicated SAM	Complicated SAM
Appetite test	Good	Poor
Generalized edema (Anasarca)	(-)	(+)
Medical complications	(-)	(+)
Management	Supervised home management.	Hospital admission.

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Home management in SAM 00:24:38

Nutrition : 175 Kcal/ kg/ day, with protein : 4 – 6 gm /kg /day.
Given by home based diet or RUTF.

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RUTF (Ready to use therapeutic food) :

Oil based paste made of

- Sugars,
- Milk solids,
- Peanut paste,
- Vegetable oil,
- In addition, added nutrients, minerals & antioxidants .

Nutritive value : 543 Kcal + 15 g protein (per 100 g)

It is stable and can be stored unrefrigerated for many months.

4 'A's

Antibiotics :

Oral Amoxicillin for 5 days.

Deworming :

Albendazole (single dose).

>2 yrs : 400 mg.

< 2 yrs : 200 mg.

Vitamin A :

- < 6 months : 50,000 IU.
- 6 – 12months : 1 lakh IU.
- > 1 year : 2 lakh IU.

Age appropriate vaccine.

Sensorineural stimulation : Emotional attachment, taking care of the child.

Home visits by health care workers (ASHA, Anganwadi, ANM).

Hospital management in SAM 00:30:14

Consists of 10 steps divided into

1. Stabilization phase (1st week in hospital) and
2. Rehabilitation phase (2nd week onwards).

Average duration of hospital stay is 6 – 8 weeks.

First 6 steps are management of complications, and the next 4 steps are feeding with stimulation.

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Active space

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	Stabilization		Rehabilitation
	Day 1 – 3	Days 4 – 7	Weeks 2 – 6
Hypoglycemia	→		
Hypothermia	→		
Dehydration	→		
Electrolytes	→	2 to 3 weeks	→
Infection	→	→	
Micronutrients	no iron		with iron →
Cautious feeding	→	→	
Catch – up growth		→	→
Sensory stimulation	→	→	→
Prepare for follow – up		→	→

Hypoglycemia :

Blood glucose < 54 mg/dl (neonates < 45 mg/dl).

Treatment :

- Asymptomatic : Give oral sugars 50 ml of 10% dextrose/ sucrose solution → regular oral feeds (breast feed in infants/ F-75 (75 Kcal/dl) every 2nd hourly in older child).
- Symptomatic(tremors,lethargy,seizures): Intravenous fluids 5 ml /kg of 10% dextrose
  (in neonate : 2 ml/kg of 10% dextrose) → 50 ml of 10% dextrose via nasogastric tube (NGT) → oral feeds.

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Hypothermia and infection must be observed as they occur as a triad, causing increased risk of mortality.

Hypothermia :

Rectal temperature < 35.5°C or axillary temperature < 35°C.

Rapid rewarming should be avoided, it may lead to Dysequilibrium syndome : Child becomes drowsy and comatose, throws seizures.

Management : Term baby – Cover the baby, Preterm : Over-head warmer, Kangaroo mother care.

Infections :

- Usual signs may not present, instead may manifest with hypothermia.
- Majority are caused by gram negative bacteria.

Active space

Leave Feedbac

- Start empirical antibiotics : Penicillin + Aminoglycosides.
- When no improvement by 48 hrs or septic shock : Add 3rd generation cephalosporin like Cefotaxime.
- Meningitis : Initial antibiotic Cefotaxime + Amikacin.

Electrolyte imbalances :

Sodium : Body sodium levels are high; serum sodium values are shown as low value or normal (diluting effect of edema) known as Dilutional hyponatremia.

Avoid excess salt or sodium in diet.

Others : K^+ & Mg^{2+} are low,

- K^+ : 3 – 4 mEq / Kg /day.
- Mg^{2+} : Day 1 : 50 % solution of MgSO4 - 0.3 ml / kg
  Day 2 : 0.8 – 1.2 mEq / Kg /day.
- Imbalances must be corrected over 2 weeks.

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Dehydration

00:46:41

Skin pinch testing is unreliable (depends on subcutaneous fat).

Signs :

- Dry oral mucosa.
- Urine output decreased (oliguria).
- Increased thirst.

It is safe to assume that all children with SAM have some dehydration.

Correction of dehydration :

Without shock :

- oral fluids, over 12 hours.
- 1st 2 hours : 5 ml / Kg every 30 min.
- Next 10 hours : 5 – 10 ml / Kg / hr.

ReSOmal (REhydration SOlution for MALnutrition) :

1 packet of WHO ORS + 2 L water + 50 g sucrose + mineral mix or 45 ml Kcl.

With shock : IV Fluid

RL with 5% dextrose or

½ NS with 5% dextrose

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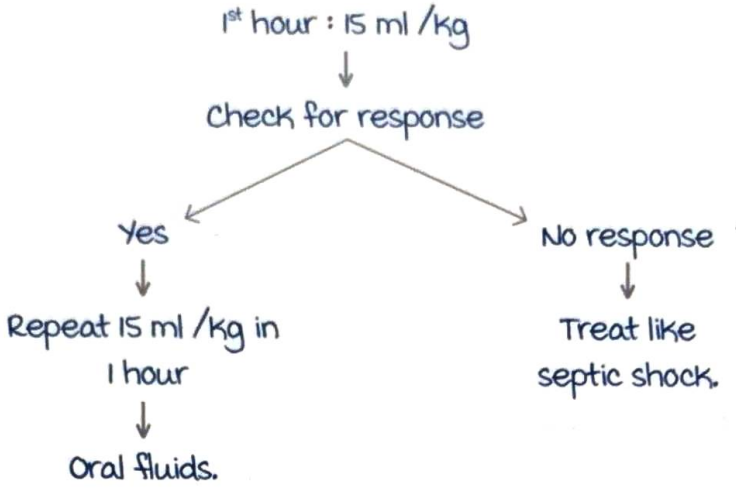

Micronutrient deficiencies :

Definite and significant micronutrient deficiency present in all children with SAM.

Recommended to give twice the RDA.

Iron is not given in the 1st week of management :

- Promotes free radical damage to intestine.
- Promotes bacterial proliferation.

From 2nd week, iron is given at 3 mg /kg /day and given for 1 – 2 months.

Vitamin A :

- If > 1 year but < 8 kg : ½ dose is given.
- If features of xerophthalmia are present :
  3 doses given : Day 0, 1, 14.

Vitamin K : 2.5 mg IM single dose.

Principle of feeding in SAM

00:58:21

In 1st week : F-75 diet is given (75 Kcal + 0.9 g protein/ 100 ml).

Initial goal : 80 Kcal/kg/day + 0.8g-1g/kg/day protein.

2nd week onwards, energy dense diet is given : F-100 diet (100 Kcal + 0.9 g protein / 100ml) → RUTF → home based diet.

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Final goal : (175 – 200 Kcal/ kg/day) + (4 – 6 g/ kg/day).

Sensory stimulation :

Structured play activity of at least 15 – 30 min / day.

Criteria for discharge :

- No edema for at least 2 weeks, and
- WFH reaches –2 S.D (or)
- MAC reaches to 12. 5 cm.

Deworming before discharge.

Active space

Leave Feedba

Treatment failure :

Primary :

Day 4 criteria :

- Failure to regain appetite.
- Failure to start loosing edema.

Day 10 criteria :

- Presence of edema.
- Failure to gain at least 5 g/ Kg/day.

Secondary :

- Failure to gain at least 5 g/ Kg/day for 3 consecutive days.

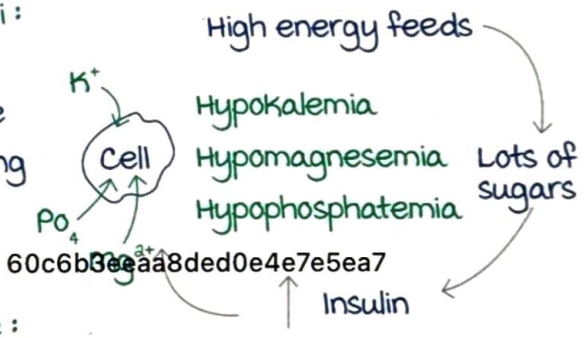
Reasons :

- Latent infection.
- Malabsorption.

Problems encountered during nutritional rehabilitation

01:06:00

Pseudotumor cerebri : Results in transient increase in ICP in the cerebrum. Self limiting so no treatment required.



Refeeding syndrome : Occurs due to calorie rich feed in the beginning rather than gradual increase.
Hypophosphatemia is characteristic abnormailty.

Nutrional recovery syndrome : Due to excess protein in the diet , associated with the development of hepatosplenomegaly.
Abdominal distension due to ascites.
Parotid swellings.
Gynaeco mastia.

Encephalitis like syndrome : Child becomes drowsy and comatose. Seen during 3 - 4 days of treatment (postulated to be due to high protein). Its very rare and self limitng.

Active space

Leave Feedbac

RICKETS AND SCURVY

These are micronutrient disorders of Vitamins D and C.

Vitamin D deficiency

00:00:25

Etiology :

- Diet : Low levels Vit D, calcium & phosphate.

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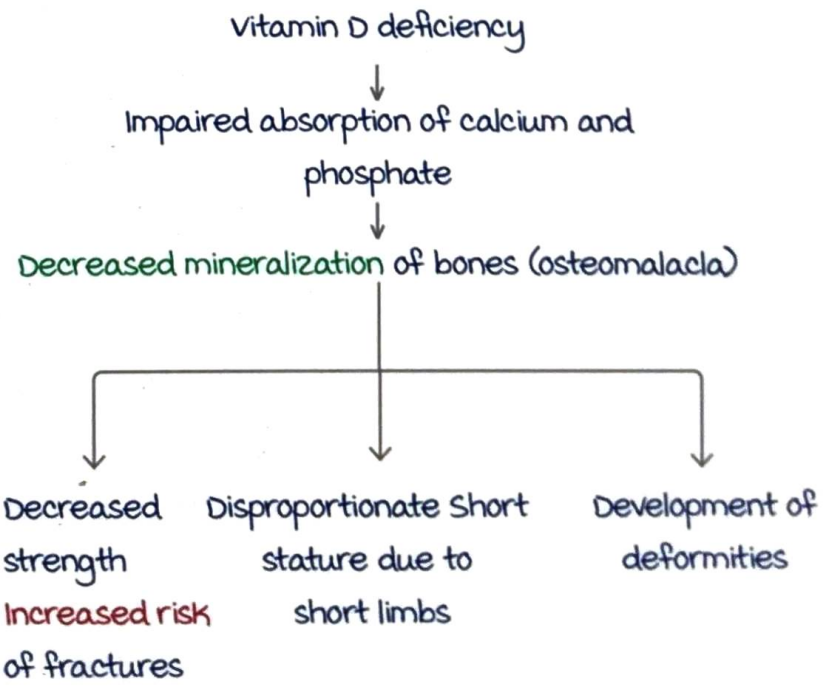
Phosphate deficiency in diet is rare except in preterm babies as they require increased phosphate for their growth.

- Sunlight : Inadequate exposure to sunlight. Vit.D is called sunshine vitamin.
- Malabsorption (fat soluble vitamins), CLD (chronic liver diease).
- Drugs : Long term use of anti convulsant, anti tubercular drugs especially Isoniazid (induces Cyt P450 → forms inactive vit D).

Pathophysiology :



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Clinical features of rickets 00:05:02

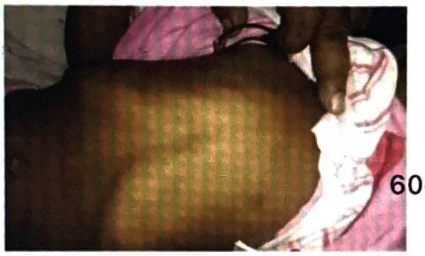
1. Deformities :

Head :

- Soft skull/craniotabes : ping pong ball consistency. It is not diagnostic. Craniotabes is also seen in hydrocephalus, congenital syphilis & osteogenesis imperfecta.
- Widely separated sutures.
- Wide open fontanelle with delayed closure.

Chest region :

Rachitic rosary : widening/enlargement of costochondral junction (string of beads appearance).



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End of metaphysis is called the growth/physeal plate.
Chondrocytes are normally present in physeal plate.
Chondrocytes
↓ After deposition of Calcium and PO_4^{3-}
Apoptosis
↓
Replaced by osteocytes → Differentiates into osteoblasts and osteoclasts.
Osteoblasts helps in forming bone/osteoid tissue.
This process is called enchondral ossification or calcification which requires phosphate. Apoptosis is inhibited due deficiency of PO_4, leading to chondrocyte hypertrophy that causes swelling in bone region.
Harrison's groove/sulcus : Due to pull of diaphragm against weak rib cage during every breath.

Limbs :

Wrist widening : Due to chondrocyte hypertrophy.
Lower limbs : Bowing of legs due to medial deviation

Active space

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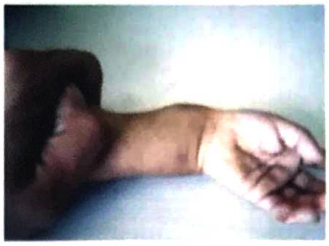
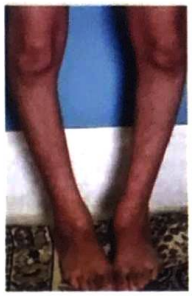
of tibia called genu varus (common in lower limb)
Knock knees : Lateral deviation of tibia (genu valgus).



Wrist widening.



Bowing of legs (Genu varus).

Knock knees (Genu valgus).

Investigations

00:15:52

1. Serum :
   - Calcium : Decreases/normal.
   - Phosphate : Decreases. Impaired absorption from intestine & increased excretion of phosphate under PTH influence.
   - Parathyroid hormone : Increases. It causes osteoclast activation leading to bone resorption. Thus, bringing calcium to normalcy.
   - Alkaline phosphatase (ALP) : Increases due to bone resorption.
   - 25 hydroxy cholecalciferol (precursor form : sensitive marker)
     himanshukitindia1@gmail.com
     (Active form : 1,25 Dihydroxycholecalciferol cannot be estimated in the lab).

2. Radiological :
   - Preferred for diagnosis.
   - X ray shows :
     1st change : Loss of normal zone of provisional calcification (ZPC). This results in irregularity at the end of metaphysis (fraying).
     Cupping (cup like shape) and splaying (widening of growth plate/ lateral deviation).

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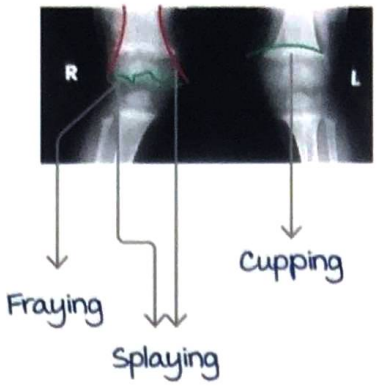

Treatment 00:20:38

Vitamin D therapy :

Oral (preferred) :

- High dose, short duration (Stoss therapy) : 60,000 IU daily or alternately for 10 days, total dose 6 lakh IU.
- Low dose, long duration (preferred) : 2000 – 6000 IU per day for 12 weeks (dependent on age).
- Maintenance : After the deficiency is corrected.
  400 IU/day for < 1 year of age.
  600 IU/day for > 1 year of age.
- Calcium 50 – 75 mg/kg/day.

Response to treatment :

- Follow up at 4 weeks.
- X ray showing healing white line of rickets due to deposition of calcium at physeal plate indicates response.
- If no response it is called refractory rickets.

Refractory rickets 00:26:56

Disorders :

1. Hypophosphatemic rickets.
2. Vitamin D dependent rickets.
3. Renal Tubular Acidosis (RTA) associated rickets.
4. Chronic Kidney Disease (CKD) associated rickets.

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X linked hypophosphatemic rickets :

- X linked dominant inheritance.
- Most commonly inherited form of rickets.
- Due to mutation in PHEX gene (Phosphate regulating gene with Homology to Endopeptidase on X chromosome).

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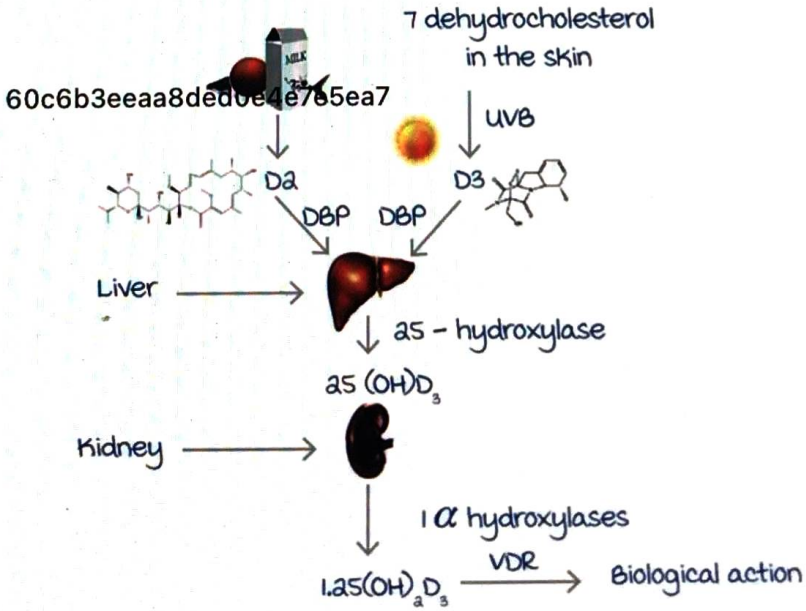
- Normal mechanism :
  PHEX gene codes → endopeptidase → inactivates phosphatonin (FGF - 23).
- Mutation :
  mutation PHEX gene → absent/reduced endopeptidase → continued activation of phosphatonin (FGF - 23) → inhibition of renal tubular reabsorption of PO_4 → PO_4 excretion in urine → hypophosphatemia.
- Presents like vitamin D deficiency rickets, but diagnosed with biochemical investigations.
- Lower limb deformities are observed.
- Dental abnormalities : Pulp defects, increased risk of dental abscess.
- Treatment :
  Oral phosphate replacement (Joulie's solution-Elemantal Phosphate).
  FGF-23 decreases active form of vit.D. Hence, Calcitriol or α calcidiol are used as supplements
  Burosumab - twaza injection (FDA approved monoclonal antibody directed against FGF - 23).

Vitamin D dependent rickets (VDDR) 00:34:37



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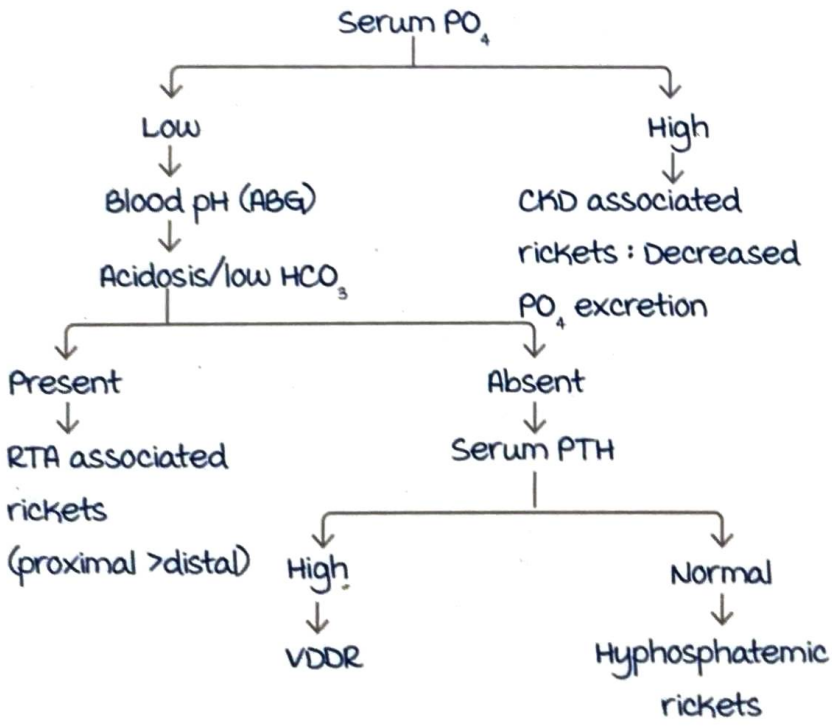
Type I VDDR	Type 2 VDDR
Congenital absence of 1-α hydroxylase. Thus, active form of vit.D is not formed. Autosomal recessive. Presents like rickets. Treatment : Calcitriol or α-Calcidiol	Resistance of Vitamin D receptors to it's action. Autosomal recessive. kumarankitindia1@gmail.com • Alopecia. • Ectodermal dysplasia/ defects : 1. Milia. 2. Presence of oligodontia (absence of >6 teeth). 3. Formation of epidermal cyst. Treatment : • Response is unsatisfactory. • High dose of calcium can give some relief.

Algorithmic approach to Refractory rickets 00:39:32



Scurvy 00:43:41

Caused by Vitamin C deficiency.

Role of Vitamin C :

- Maintenance of connective tissue (collagen).

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Collagen is formed by hydroxylation of proline & lysine.
Vitamin C is needed for the process of hydroxylation.

- Wound healing (similar to zinc).
- Absorption of iron from intestine.
- Osteoid formation.

Vitamin C rich foods : Citrus fruits (orange & lemon) and green vegetables.

Risk group :

Children consuming only pasteurized cow milk (breast milk has sufficient Vitamin C).

Children consuming diet lacking vitamin C.

Features of scurvy :

Early features :

1. Irritability.
2. Decreased appetite.
3. Anemia.
4. Petechiae (increased fragility of capillaries).
5. Bleeding manifestations :
   - Gums.
   - Perifollicular bleeds : Corkscrew appearance of hair.
   - Subperiosteal bleed : Pseudo paralysis due to painful movement (frog leg posture- flexion at hip and knee).

Bleeding gums

6. Scorbutic rosary :
   - Tenderness.
   - Angulated appearance.

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Scorbutic rosary (Vit. C deficiency)	Rachitic rosary (Vit.D deficiency)
Tenderness +	No pain
Angulated rosary	Rounded rosary

Radiological findings in scurvy 00:50:15

On X ray :

1. White line of Frankel (well calcified cartilage).
2. Scorbutic/Trummerfeld's zone (Central zone of rarefaction) : Represents poorly formed bony trabeculae.
3. Pelken's spur : Lateral zone of rarefaction.
4. Ring epiphysis/Wimberger's ring.
5. Ground glass appearance.
6. Pencil thin cortex.

Diagnosis is made by clinical appearance of the child.

Investigations :

Leukocyte vitamin C levels using Buffy coat.

Serum vitamin C level (best).

Treatment :

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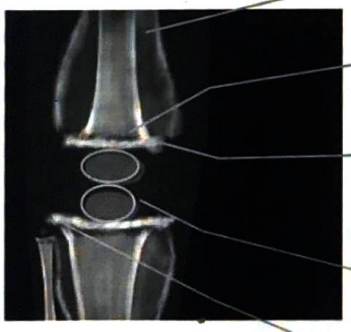
100 to 200 mg/day of vitamin C (equivalent to 100 ml of orange juice or tomato pulp).



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GENETIC DISORDERS

Aneuploidies :

The difference in the normal number of chromosomes :

- Trisomy : An extra chromosome makes a total of 3 instead of a pair. E.g. Trisomy 21.
- Monosomy : Only one chromosome instead of a pair. E.g. Monosomy X : only one x chromosome in a female.

Down syndrome - Trisomy 21 00:01:28

It is the most common trisomy. Most common trisomy in live babies & m/c genetic cause of low IQ/intellectual disability.

Genetic basis of Down syndrome :

- Maternal meiotic non disjunction of chromosome 21 is the most common cause. Seen in 95% of cases.
- Robertsonian translocation : Refers to translocation between two acrocentric chromosomes. Seen in 4% of cases. E.g. t (14,21), t (21,21).
- Mosaicism : A mixture of both trisomy 21 cells and normal cells in a child. Seen in 1% of cases.

External characters : Very characteristic.

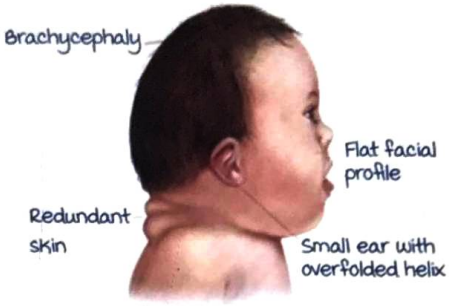
Dysmorphic facies :

- Mongoloid slant of eyes : Eyes are oriented upwards. (Upslanting of palpebral fissure).
- Brushfield's spots : White spots on iris.
- Epicanthal folds : Extra fold of skin that increases the space between eyes and nose.

Active space

Lateral view :

- Brachycephalic skull with a flat nose, occiput & face.
- Low set ears : The entire ear is below the imaginary line drawn horizontally from the outer canthus (normally part of the ear will be above the line).
- Protruding tongue due to the hypoplastic mandible.



Hands :

- Simian crease : single transverse palmar crease (normally 2 creases are present).
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- Clinodactyly : The little finger is curved.

Feet :

- Sandal gap : Wide gap between first and second toe.

Internal defects/anomalies :

- Low IQ and hypotonia.
- Cardiac defects :
  - They are the most common cause of mortality and morbidity in children with Down syndrome.
  - The heart is the most common organ affected. seen in 50% of children with Down syndrome.
  - Common order is : Endocardial cushion defect/atrioventricular septal defect > VSD > ASD.
- Gastrointestinal defects seen are :
  - Most common : Duodenal atresia.
  - Hirschsprung disease.
  - Annular pancreas.
- Increased risk of hypothyroidism is seen in 30% of cases. Hence, regular screening is to be done.
- Increased risk of cancers :
  - ALL : Overall most common leukemia.
  - AML (M7 : megakaryocytic type) is the most common in children less than 3yrs.

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common in children <3 years of age.

Pre-senile Alzheimer's disease is seen : Onset at 40 years.

- Chromosome 21 has a specific gene that codes for APP (Amyloid Precursor Protein).
- Extra chromosome 21 : Results in increased production of beta amyloid. It builds up in the brain & predisposes to Presenile Alzheimer's disease.

Atlanto axial subluxation is seen with short neck. Hence screening neck X ray by 3-5 years of age is to be done. These children are advised to avoid contact sport activities.

Antenatal screening tests 00:15:43

1. Antenatal USG :
   - The marker is increase in nuchal translucency (1st trimester) or thickness (2nd trimester) : ≥ 3 mm is suggestive.

   Other clues suggestive of Down syndrome :
   - Hypoplastic or absent nasal bone.
   - Short femur.
   - Echogenic focus in the heart.

2. markers from maternal blood :
   - In 1st trimester, double test is done which includes Pregnancy Associated Plasma Protein - A (PAPP - A) and β HCG.
   - In the 2nd trimester, triple test is done which includes β HCG, alpha feto protein and unconjugated estriol.
   - Quadruple test : Triple test + inhibin A.

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Interpretation of markers :

- β HCG & inhibin A are elevated.
- Other markers are reduced.

Generally, in pregnancy, a combination is used :

- Combined test : Includes 1st trimester USG + double test.
- Integrated test : In 2nd trimester, combined test + quadruple test.

Active space

3. Cell Free Fetal DNA testing (CFF DNA testing) : Some fetal DNA enters maternal circulation and can be detected in blood. It is also called Non Invasive Prenatal Test (NIPT).

Confirmatory test : Karyotyping 00:22:28

The sample is obtained only by invasive methods :

- CVS : Chorionic villous sampling at 11 to 13 weeks.
- Amniocentesis is done around 15 weeks of gestation.

A confirmatory test is done only after screening test showing high risk , due to possibility of inducing abortion or termination by invasive tests.

Recurrence risk of Down syndrome :

- General risk : 1 %.
- Advanced maternal age (> 35 years) : 4 %.
- Robertsonian translocation in parents :
  - t (14,21) in father : 3 to 5 % (< 5%).
  - t (14,21) in mother : 10 to 15 %.
  - t (21,21) in father or mother : Risk is 100%.

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Other trisomies :

- Trisomy 16 : Most commonly associated with spontaneous abortion.
- Trisomy 18/Edward syndrome : 2nd MC trisomy.
- Trisomy 13 : Patau syndrome.

Edward syndrome (Trisomy 18) 00:28:09

External characters :

- Prominent occiput.
- Microphthalmia (short and tight palpebral fissures).
- Increased risk of cleft lip/palate (more with patau syndrome).
- Closed fists with overlapping of fingers.
- Short neck.
- Rocker bottom foot : Prominent heels. The appearance of the foot resembles the bottom of a rocking chair.

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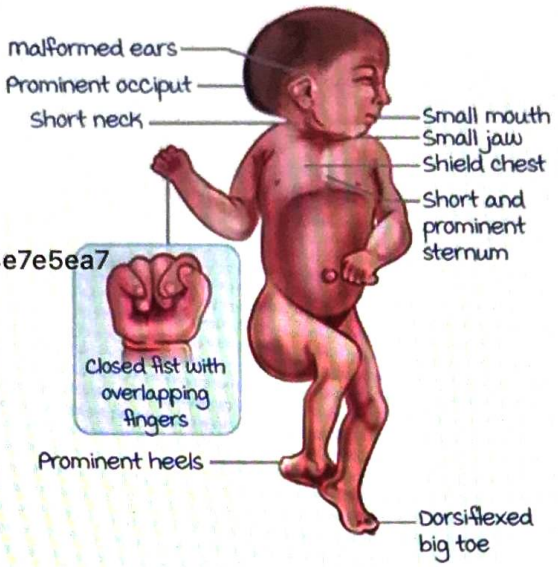
- Limited hip abduction.



Rocking chair

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Associated organ defects or anomalies :

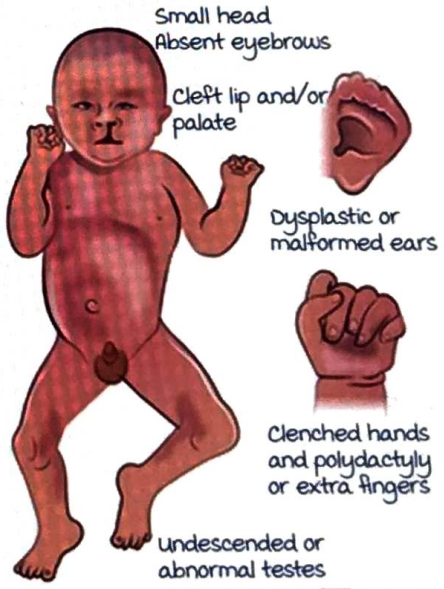
- Cardiac defects : > 90% of cases.
- VSD > PDA > ASD.
- Renal defects (Horseshoe kidneys)

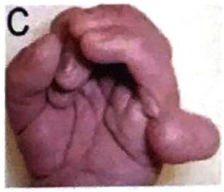
Patau syndrome (Trisomy 13)

00:32:37

External characters :

- Cleft lip or palate (60 to 80 % cases).
- Microphthalmia.
- Dysplastic or malformed ears.
- Aplasia cutis : Absence of skin in areas of scalp
- Fingers : Postaxial polydactyly. Clinodactyly.
- Undescended testis : Cryptorchidism.





Polydactyly

Organ defects :

- Heart defects : (80 to 90% cases). VSD > PDA > ASD.
- Absent or missing ribs.
- Holoprosencephaly or arrhinencephaly.

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Monosomy X : Turner syndrome

00:36:58

It is represented as 45 XO.

O for the absent X chromosome.

Genetic basis : Due to loss of X chromosome during paternal gametogenesis.

Among the cases, after Karyotype analysis :

- 57% have Complete monosomy X.
- 29% have Mosaicism.
- 14% have Partial monosomy X. Other X chromosome is not completely deleted, it is a ring chromosome or isochromosome.

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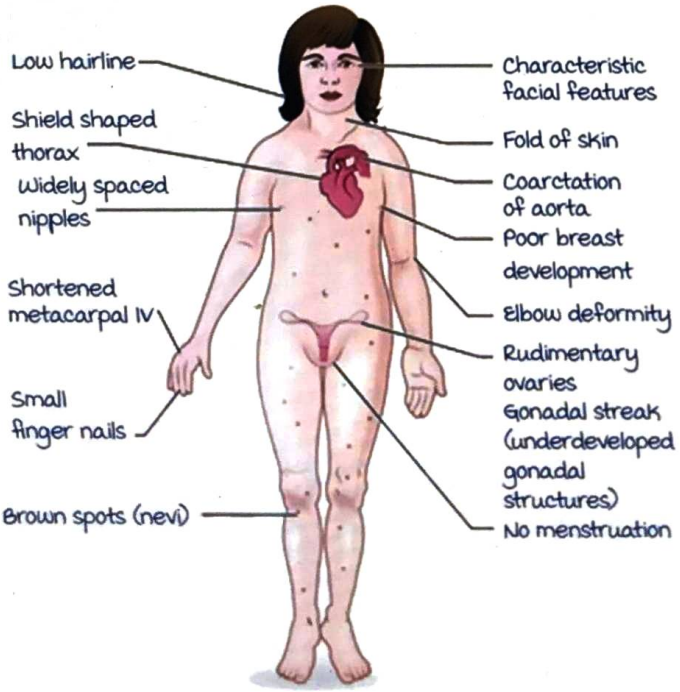
External features :

- Short stature (< 145 cm even as adults).
- Triangular face.
- Neck : Loose skin folds.
  Webbed neck.
  Low posterior hairline.
- Chest : Flat (shield) chest & widely spaced nipples.
- Increased carrying angle > 135° i.e. cubitus valgus.
- Short 4th metacarpals.



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Increased risk of organ defects :

- Heart : Bicuspid aortic valve > coarctation of the aorta.
- Gonads : Underdeveloped ovaries (streak ovaries), reduced oestrogen production resulting in infertility & amenorrhea.

Presentation in neonatal period or infancy :

- Dorsal lymphedema of feet & hands, due to lymphatic obstruction.
- In the neck, it is called : Cystic hygroma.

Normal IQ but they have learning disabilities.
Turner Karyotype 45 XO is the m/c chromosomal abnormality seen in spontaneous abortions.
m/c trisomy associated with spontaneous abortions in pregnancy is trisomy 16.

Management :
Estrogen replacement till they acquire secondary sexual characters + recombinant growth hormone (rGH) to attain height.

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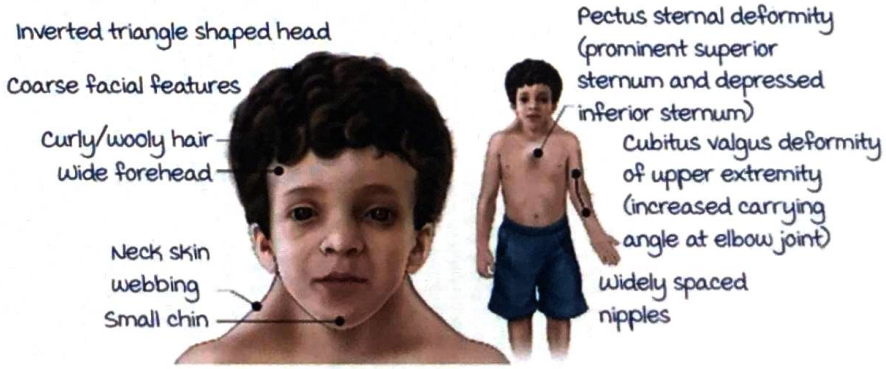
Noonan syndrome/pseudo turner syndrome 00:46:19

Not an aneuploidy. Seen in females as well as males.
Features are similar to Turner syndrome.
Autosomal dominant chromosome 12q24.1
PTPN gene : Protein Tyrosine Phosphatase Non receptor.



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Features like Turner syndrome are :

- Triangular face due to small chin.
- Flat chest.
- Widely spaced nipples.
- Cubitus valgus.
- Webbing of the neck.

Differences between Turner and Noonan syndrome :

	Turner	Noonan
Karyotype	45 XO (female only).	46 XX (female), 46 XY (male).
IQ	Normal.	Low IQ.
Gonadal defects	Streak ovaries.	Cryptorchidism (low testosterone. Delayed puberty and low fertility)
Cardiac defects	Bicuspid aortic valve. Coarctation of aorta.	Pulmonary stenosis. Hypertrophic obstructive cardiomyopathy (HOCM). Atrial septal defect.

Klinefelter syndrome

00:50:48

- Karyotypic abnormality : 47 XXY.
- Most common sex chromosomal aneuploidy.
- Due to non disjunction of the X chromosome during meiosis of gametogenesis, in either parent.
- Hence, paternal & maternal defects equally contribute to this condition.
- Risk factor : Increase in paternal age.
- Klinefelter syndrome is the most common genetic cause of infertility/hypogonadism in males.

Clinical features :

- Taller than average height (legs are unusually long).
- Cryptorchidism leads to low levels of testosterone.
- Eunuchoid body habitus : Absence of axillary & facial hair. (Delayed appearance of secondary sexual characters).

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Active space

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- Small penis and testis, Atrophy of testis & seminiferous tubules at late stages. Increased incidence of infertility.

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Puberty in Klinefelter's syndrome :

- It starts at a normal age.
- But the progression is very delayed.

Gynecomastia : Due to increased activity of enzyme aromatase, it converts testosterone into estrogenic metabolites (estradiol).
An increase in estrogen metabolites increases the risk of male breast cancer.

Low IQ, learning disabilities (verbal skills affected).
Behavioural problems.

Increased risk of extragonadal germ cell tumors, especially in the anterior mediastinum.

Microdeletion syndromes

00:59:26

- Small deletions which are not detectable in Karyotyping.
- FISH (Fluorescent In Situ Hybridization) : For diagnosis.

Di George Syndrome :
22q11.2 microdeletion.
Abnormalities in the development of 3rd & 4th pharyngeal pouches.
The thymus & parathyroid are affected.

Mnemonic for clinical features : CATCH 22.

Cardiac : Conotruncal defects.

- Tetralogy of Fallot.
- Interrupted aortic arch.
- Right sided aortic arch.

Abnormal facies.

- Hypertelorism : widely separated eyes.
- Thin upper lip/small mouth.

Active space

- Small jaw.
- Low lying & notching of ears.

- T cell deficiency due to thymic hypoplasia/aplasia.
- Cleft lip/palate.
- Hypocalcemia due to low parathyroid hormone.
  It can result in seizures during infancy.

DiGeorge syndrome is also called Velocardiofacial syndrome : Velum (palate), cardiac & facial abnormalities.

William syndrome

01:10:04

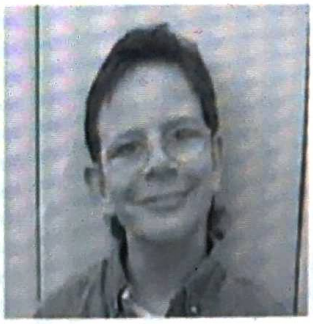
Microdeletion in 7q11.23

Facial appearance :

- Wide mouth.
- Prominent forehead.
- Puffiness in eyes & cheeks.

These features resemble elves : Elfin facies.

These children are happy & friendly personalities.



Associated problems :

- Hypercalcemia.
- Cardiac defect : Supravalvular aortic stenosis.
- Low IQ, learning disabilities.

Genomic imprinting disorders

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01:09:56

Each gene has 2 alleles paternal and maternal. Out of them, one is normally silenced or imprinted and only one is active.

In this disorder :

- Deletion of active allele (most common) or
- Disomy of inactive allele.

Active space

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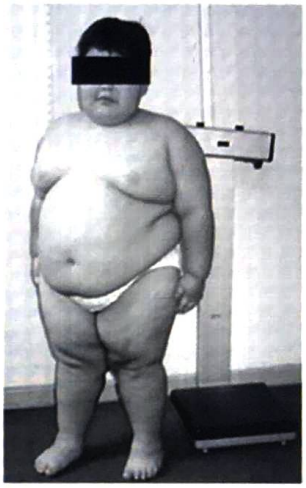
Examples :

- Prader Willi syndrome.
- Angelman Syndrome.

Prader willi syndrome

01:09:56

Deletion of the paternal allele in chromosome 15q (15q11–13) is the common cause (or)
Disomy of the maternal allele.



Clinical features :

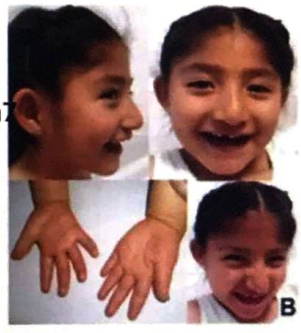
- Obesity : Increased intake of food (hyperphagia) due to an increase in the hormone ghrelin which stimulates the appetite.
- Shorter compared to children of the same age.
- Small almond shaped eyes, small hands & feet.
- Hypogonadism : Gonads are not properly developed, resulting in delayed puberty.
- Low IQ.

Angelman syndrome

01:13:20

In chromosome 15q11–12.

- Deletion of the active maternal allele is the common cause (or)
- Disomy of paternal allele.



Clinical features :

- Happy puppet syndrome : Inappropriate laughter + ataxia (involuntary movement of hands and feet).
- Very low IQ.
- Increased incidence of seizures.
- Hypotonia.
- Feeding difficulties.

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Other genetic syndromes :

Treacher collins syndrome

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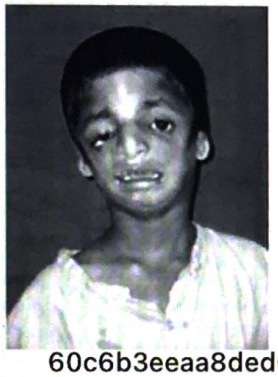
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- Autosomal dominant disorder.
- Also known as mandibulofacial dysostosis.

Clinical features :

- Slant of eyes : Sloping downwards (antimongoloid slant).
- Coloboma of lower eyelids.
- Sunken cheeks.
- Under developed or small mandible.
- Dental malocclusion (malaligned teeth).
- Abnormal hair distribution.
- Pre auricular hair displacement.
- Increased risk of conductive hearing loss due to malformation of ossicles and defective development of the middle ear cavity.
- Intelligence is normal.

Pierre robin sequence

01:20:20

It is not a condition by itself but is associated with other conditions.

It is a sequence or chain of events.

The order of the sequence :

1. Severely under developed mandible in utero (common starting point)
2. The tongue is pushed back : Retroglossoptosis.
   It Interferes with the development of palate, resulting in a big 'U shaped' cleft palate that corresponds to the shape of the tongue.

Can predispose to aspiration of feeds after birth.

3. Results in airway obstruction after birth because of toungue being pushed back.

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Associations with other syndromes :

- Fetal alcohol syndrome.
- Trisomy : Trisomy 13.
- In Treacher Collins syndrome, severely affected mandible results in Pierre robin sequence.

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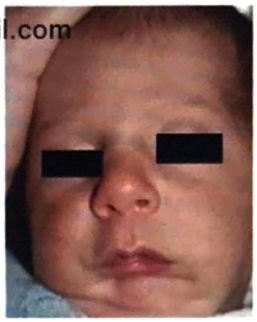

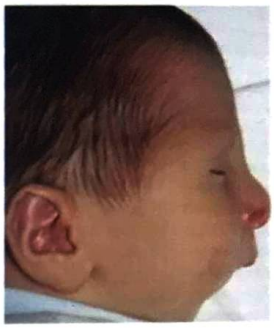

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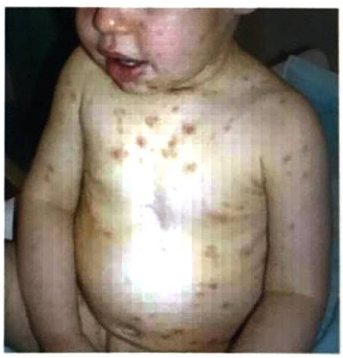
COMMON CHILDHOOD INFECTIONS

Mnemonic for day of appearance of rash in febrile child :
Very Sick Persons Must Take Rest.

Varicella	1st day.
Scarlet fever	2nd day.
Pox (small)	3rd day.
Measles	4th day.
Typhus (Epidemic)	5th day.
Rickettsia	6th day.

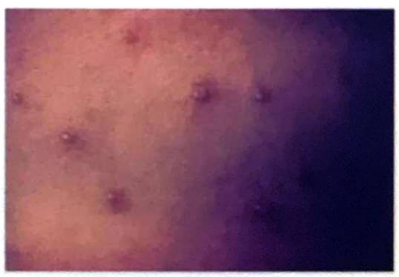
Varicella : Chicken Pox 60c6b3eeaa8ded0e4e7e5ea7 00:02:41

Causative agent : Herpes Zoster Virus (double-stranded DNA virus).
Incubation period : 10 to 21 days.
Period of Infectivity : 24 to 48 hours before the onset of rash till all the lesions are crusted.
Highly infective with a secondary attack rate for household contacts : 80%



Features :

Fever.
Rash :
On 1st day of fever.
Initially macular rash turns to papular and to vesicular rash successively.
The characteristic vesicular rash is intensively pruritic.



Centripetal rash : Starts in the trunk, and spreads to limbs.

Active space

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Rash lasts for 3 to 5 days and leaves behind a brown pigmentation when it goes away.

Pleomorphic rash : Different sizes of rash are seen depending on the different stages in the evolution of rash.

Dewdrop in rose petal appearance : Rash is a clear fluid filled vesicle surrounded by erythema.

Complications :

- Secondary bacterial infection by Staph. aureus or Strep. pyogenes.
- Neurological : Meningoencephalitis, Cerebellar Ataxia.
- Risk of Reye's Syndrome : Avoid Aspirin.

Varicella : Diagnosis

00:09:14

Is based on clinical presentation.
Tzanck smear examination : Scraping of lesions.
Anti IgM antibody demonstration against varicella virus.

Treatment :

- Symptomatic treatment : Antipyretics (like Paracetamol).
- Avoid Ibuprofen : Risk of necrotizing fasciitis.
- Acyclovir : Antiviral of choice dose : 20 mg/kg/dose every 6th hrly.
  To be started within 48 hrs. Not of much benefit later.

Period of isolation :
The child must not attend school until no new lesion appears and all existing lesions have crusted.

Prevention :

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Vaccine strain : Oka strain, live attenuated.
Dose : 0.5 ml S/C.
Schedule :

- 1st dose : 15 to 18 months of age
- 2nd dose : 4 to 6 years of age.

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Post-exposure prophylaxis :

Varicella-Zoster Immunoglobulin.

Recommended only to high-risk individuals :

Pregnant women.

To neonate whose mother developed chickenpox within 5 days before to 2 days after delivery.

Immunocompromised patients.

Scarlet fever

00:14:29

Etiology : Group A β hemolytic streptococci.

Age group : School going children 5 yrs to 15 yrs.

Features :

Fever with rash on 2nd day.

Characters of rash :

- Starts from face.
- Fine maculopapular rash.
- Spreads to trunks.

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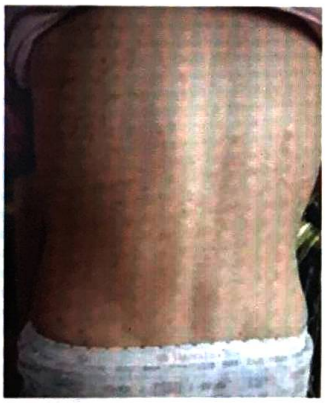

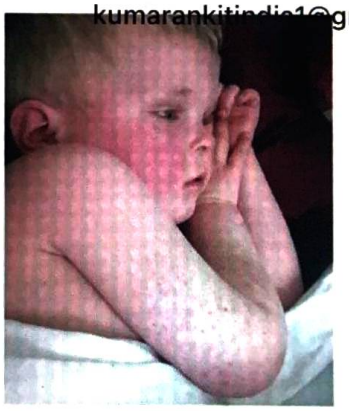

Never involves palms & soles.

Sandpaper rash : As the rash makes the skin appears rough.

Pastia's lines : Accentuation of rash in the skin crease.

Other findings :

- Sore throat : Pus in the throat.
- Associated tender cervical lymphadenopathy.
- Strawberry tongue : Due to intense inflammation.
  Hypertrophy of papillae.

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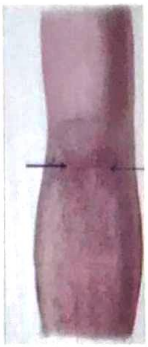


Investigation :

Culture of throat swab : Investigation of choice.

Treatment :

Penicillin G for 10 days is the treatment of choice.

Complications :

Post streptococcal complications :

- Acute rheumatic fever.
- Post streptococcal glomerulonephritis.
- Post streptococcal reactive arthritis.

Measles

00:20:12

Viral infection : Single-stranded RNA virus of family Paramyxoviridae.

Incubation period : 8 to 12 days.

Secondary attack rate : more than 90%. Highly infective.

Infectivity period : 4 days before the appearance of rash to 5 days after the appearance of rash.

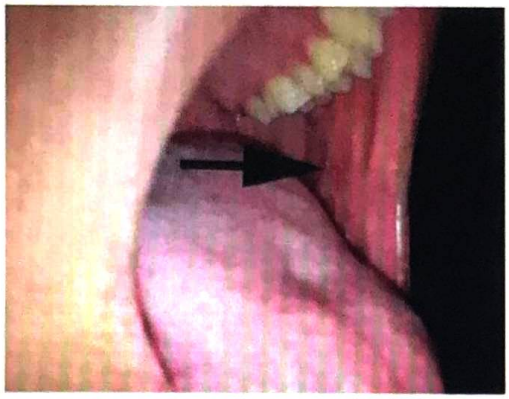
Characteristic features :

Initially by day 1 to 3 fever and prodromal features (cough, conyza, conjunctivitis).

Between days 2 to 3 : Koplik spots (pathognomic of measles).

- They are white spots surrounded by redness.
- Rice grain appearance surrounded by redness or erythema.
- Location : Buccal mucosa : Opposite to second lower molar.

Active space



Koplik spots

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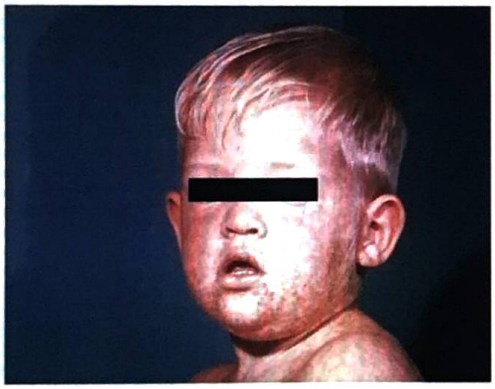
Day 4 : Rash appears.
Maculopapular rash.
It starts from the face : Behind the ear.
Spreads to trunk and then to extremities.
The duration of rash is 4 days, it disappears leaving brownish hyperpigmentation.

On day 8 : Rash disappears.



Maculopapular rash

Complications :
Most common acute complication : Otitis media.
Most severe complication : Bronchopneumonia.

- Gaint cell pneumonia/Hecht's pneumonia : Etiological agent is the virus itself.
- Secondary bacterial infection : Due to Strep. Aureus and Staph. Pneumonia.

Active space

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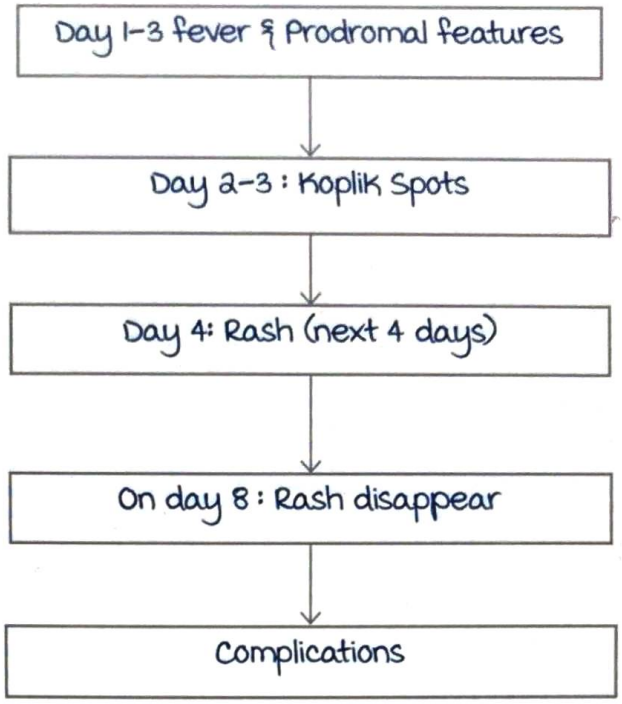

Subacute Sclerosing Pan Encephalitis - SSPE

00:27:18

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Chronic complication :

occurs 7 to 13 years after infection.

Decrease in school performance.

Behavioural problems.

myoclonic seizures.

A fatal condition.

Investigations :

- CSF testing to look for measles specific antibodies : IgG antibodies.
- EEG : Rademecker complexes : Periodic high voltage discharges on a slow background.

Treatment :

- Isoprenosine along with Interferon gamma (by intrathecal route).

Measles : Treatment :

Supportive only : IV/oral fluids.

Vitamin A : Single-dose (orally) to be given : Reduces mortality and morbidity.

Active space

- Less than 1 year : 1 lakh IU.
- More than 1 year : 2 lakh IU.

Prevention by vaccine :

MR vaccine : Measles-Rubella vaccine.

Is included in the National Immunisation Schedule.

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00:32:36

Exanthema subitum

On the 4th day of fever, the rash appears.

Also called as sixth disease or Roseola infantum.

Etiology : Human Herpes virus (HHV) 6/7, Echovirus type 16.

MC : HHV 6.

Features :

Fever + Viral prodrome : Lethargy, cough and running nose.

The rash (maculopapular rash) appears on the 4th day. It has to be differentiated from measles.

- Rash is maculopapular.
- Starts from the trunk region.
- After the rash starts fever will decrease.
- No residual pigmentation after the rash disappears.

Nagayama spots : Ulcers at uvulopalatoglossal junction.

Treatment :

Symptomatic treatment and the prognosis is excellent, not associated with any complications.

Active space

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Rubella

00:37:50

Etiology : Single-stranded RNA virus : Togaviridae.
Incubation period : 14 to 21 days.
Infectivity period : 5 days before to 6 days after the Rash.
Other names : 3 days measles/German measles.

Features :
The rash appears to be similar to measles : maculopapular and starts from the face.
The total duration of the rash is 3 days.
Prodromal features : Before the rash is either present or absent.
1st sign is usually rash.

- maculopapular.
- Starts from the face and spreads to the trunk.
- Duration is 3 days.
- No residual pigmentation or desquamation after the rash disappears.

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Rubella other features :
Forchheimer spots : In oropharynx – Tiny, rose lesions.
Lymphadenopathy : Suboccipital, postauricular and anterior cervical lymph nodes.

Rubella : Complications rarely.

- Post thrombocytopenia : 2 weeks after the rash.
- Arthritis : Adolescents/adults.
- Most severe : Progressive rubella panencephalitis : Fatal disease.

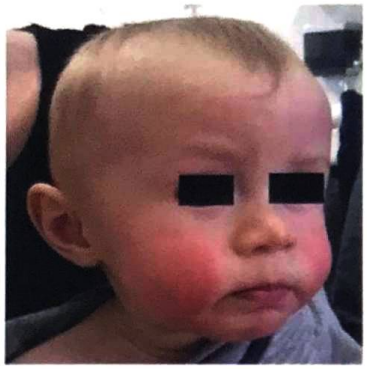
Prevention : MR vaccine.

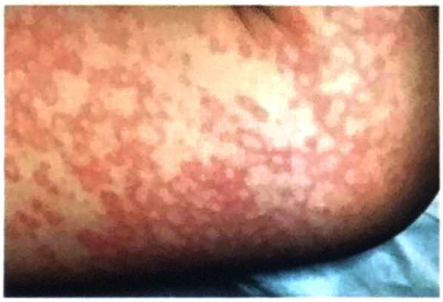
Active space

Erythema infectiosum

00:43:48

Etiology : Parvovirus B19, single-stranded DNA virus.
Incubation period : 4 to 28 days.
Age group : School going children.





Features :

Fever with prodrome : 1 to 2 days,

Rash :

- Slapped cheek appearance : Intense redness in cheeks.
- Starts at the face and spreads to the trunk.
- Central clearing is seen in rash : Lacy or reticulated appearance.

Other associations :

- Arthralgia/arthropathy : Older children.
- In children with chronic haemolytic anaemia : It can predispose them to transient aplastic crisis, here pancytopenia is seen.
- If the condition occurs in pregnancy can lead to hydrops fetalis and can be associated with intrauterine death or stillbirth.

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Treatment : Self-limiting, supportive measures only.

Epidemic typhus

00:47:54

Causative agent : Rickettsia prowazekii.

Transmission : Faeces of infected body louse.

Features :

Triad : Fever, rash, headache.

It is seen in 50 % of patients.

Rash on day 5 of fever.

Purpuric rash involves palms & soles.

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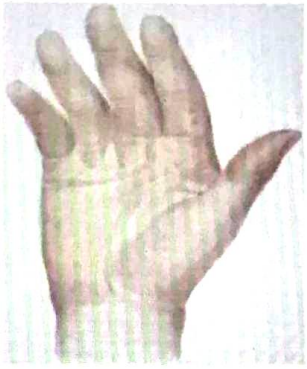


Drug of choice : Doxycycline for 5 to 7 days and for at least 3 days after the patient becomes afebrile to prevent relapse.

Hand foot mouth disease (HFMD) 00:51:03

Common viral illness in children < 5 years.
Usually seen as outbreaks in preschools.
Causarive agent : Coxsakie A16 and enterovirus 71.

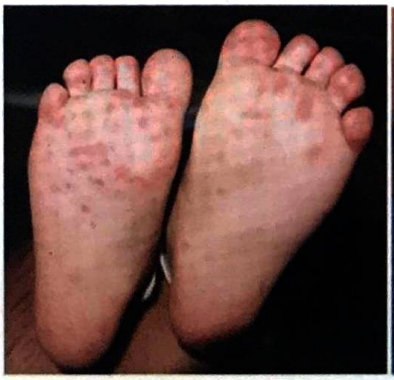
Transmission :
Direct contact or usage of infected fomites.

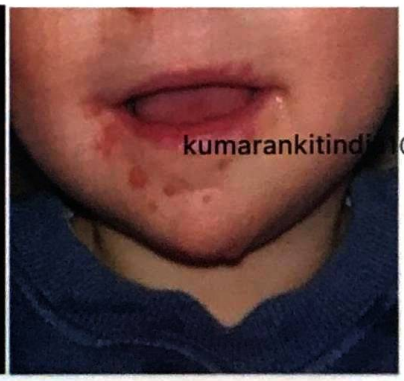
Features :

- Fever + ulcers or blisters in oral cavity.
- Classical blisters are seen around the mouth.
- In palms and feet : Papulovesicular lesions are seen.
- Lesions resolve by 4 to 5 days.

Does not usually cause complications.

Treatment is symptomatic with Isolation of affected children at home till all lesions are resolved.





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Active space

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TORCH INFECTIONS

Intra uterine infections 00:00:16

Transmitted from mother to baby in intra uterine life.

Congenital Infections.

Toxoplasmosis.

Others (Syphilis, Zika virus).

Rubella.

Cytomegalovirus (most common).

Herpes (Herpes simplex, Herpes zoster).

Clinical features :

- Primary infection of mother that affects the baby (except in syphilis, even latent infection can affect the baby).
- Most transmissions occur in the first trimester of pregnancy.
- Risk of congenital anomalies in the baby is high only if infection occurs in the first trimester (organogenesis).

Common manifestations of Intrauterine infections :

Antenatal :

- Intrauterine growth restriction (m/c).
- Intrauterine death.
- Preterm baby.
- Abortions.
- Recurrent abortions (Syphilis).

Post natal :

Only if baby is severely affected :

- Anemia.
- Thrombocytopenia → Petechiae.
- Hepatosplenomegaly.
- Jaundice (abnormal LFTs).

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Congenital Rubella syndrome 00:06:17

Most severe intrauterine infection.

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Triad : Cataract + deafness (sensorineural hearing loss) + cardiac defects.

- Heart defects :

  PDA > Pulmonary artery stenosis (R > L) > VSD.

MC ocular manifestation : Salt and pepper retinopathy.

MC manifestation : Deafness (sensorineural hearing loss).

Most distinctive feature : Chronicity.

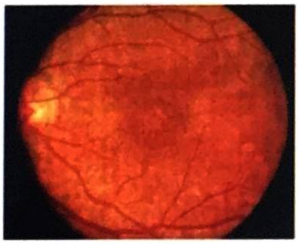
- Virus survives for a long time in brain → Progressive rubella panencephalitis (PRP).

  Features of PRP :

  Seizures.

  Deteriorating school function.

  Intellectual impairement.



Salt and pepper retinopathy.

Other features :

- Increased risk of diabetes.
- Increased risk of hypothyroidism.

Diagnosis :

- Demonstration of IgM rubella antibodies (can repeat after a few days of suspicion high, but test negative).
- Throat swab → Culture/PCR.

No treatment.

Vaccination : Live attenuated vaccine (RA 27/3 strain) (MMR vaccine).

Congenital Cytomegalovirus infection 00:12:06

MC TORCH infection.

Risk of transmission of CMV from mother to baby is high in the 3rd trimester.

Transmission :

1. Congenital/intrauterine transmission :
   - 3rd trimester.
   - Around 90% babies asymptomatic.

Active space

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- < 5% babies affected → Congenital cytomegalic inclusion disease.

Triad : Microcephaly + chorioretinitis + intracranial calcifications.

Calcifications : Periventricular/around the ventricles.

MC long term sequealae : Sensorineural hearing loss.

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Diagnosis :

- PCR testing (test to be done within 2 weeks after birth as 2 weeks intrauterine and post natal infections cant be distinguished).
- Samples used : Urine (best) > blood > saliva (screening).
- Igm test.

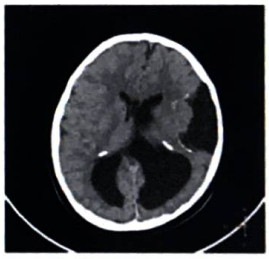
Treatment :

DOC : Ganciclovir (only if baby has progressive neurological disease or sensorineural hearing loss).

IV ganciclovir for first 6 weeks → Oral ganciclovir 6 months.

2. Post natal transmission :

- More common.
- Transmission through breast milk.
- Baby usually asymptomatic.
- Infected babies are reservoirs.



Intracranial calcifications

Preterm/very low birth weight baby : Presents like a sepsis like illness or pneumonia with respiratory distress.

Congenital Toxoplasmosis 00:19:38

- Risk of transmission high in the 3rd trimester.
- Mostly asymptomatic.
- If infection occurs in 1st trimester → Baby presents with following clinical features.

Triad : Chorioretinitis + intracranial calcifications + hydrocephalus.

Calcifications : Diffuse intracranial calcifications (scattered throughout the brain).

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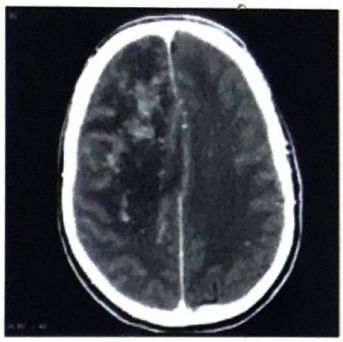
Diagnosis :

Serology (Double sandwich ELISA testing : IgM/IgA ELISA).

- IgA ELISA : more sensitive.
- IgM ISAGA (Immuno Sorbent Agglutination Assay).

Treatment :

- All babies should be treated.
- Pyrimethamine + Sulfadiazine for 1 year.
- Add supplementation : Folinic acid/Leucovorin.



Diffuse intracranial calcifications

Prevention : Fruits and vegetables should be washed, pregnant woman should avoid eating uncooked food.
If mother already affected, she can be given Spiramycin (decreases transmission risk).

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Herpes zoster/Varicella infection

00:25:45

Modes of transmission :

1. Perinatal :
   - more common, less severe.
   - At risk period : When mother gets infection just 5 days before/2 days after delivery.
   - Administer varicella zoster immunoglobulin or IV Ig to the baby after birth to prevent lesions.
   - If lesions developed → Acyclovir.

2. Intrauterine or congenital :
   - more severe.
   - Risk of transmission is more in the 2nd trimester.

   Manifestations :
   - Cerebral atrophy → Microcephaly.
   - Hypoplasia of limbs (short limbs).
   - Scars (similar to burns) : Cicatricial scarring (zig zag).

Active space

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Herpes simplex infection :

Usually transmitted in the perinatal period (when baby comes in contact with the vesicles during delivery).

Therefore, cesarean section (LSCS) is more preferred in affected mothers.

HSV 2 is more common.

3 patterns of presentation :

1. Classical pattern (MC) : Development of vesicles in skin, eye and mouth.
2. CNS manifestations : meningoencephalitis with seizures.
3. Disseminated pattern : Similar to sepsis.

Not associated with vesicles

Diagnosis :

- PCR testing.
- Tzanck smear from lesions.

Treatment : Acyclovir for 6 months.

Congenital Syphilis

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00:32:30

- Rare.
- Only intrauterine infection which is associated with recurrent abortions.
- Usually transmitted after 4 months of gestation (2nd and 3rd trimesters).

Lesions :

Early onset (within 2 years after birth) :

- Vesiculo bullous rash (can involve hands & feet also).
- X rays : Intense inflammation of bone (periosteitis) or osteochondritis or metaphysitis.
- Intense Inflammation in limbs → Severe pain → Pseudo paralysis of Parrot (child doesn't move limbs).
- Inflammation of oral and nasal mucosa → Continuous discharge from nose and mouth (similar to rhinitis) called snuffles.

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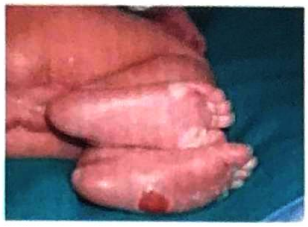
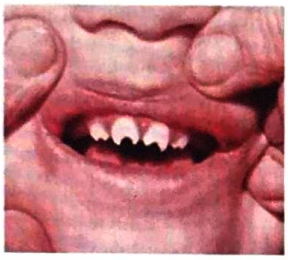
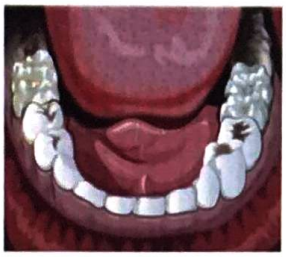
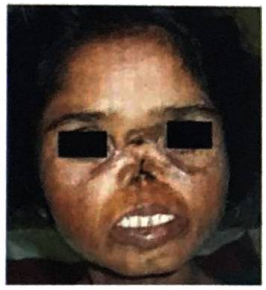




Hutchinson's teeth



Mulberry molars in syphilis



Deformed face

Late onset (after 2 years of birth) :

- Hutchinson's triad : Hutchinson's teeth (notching/ peg shaped incisors) + interstitial keratitis + deafness (sensorineural hearing loss).
- Mulberry molars : Irregular surface (not a part of Hutchinson's triad).
- Deformed nose (intense inflammation → Destruction of nasal septum and bone).
  Initially : Saddle nose deformity.
  Later : Complete destruction of nose.
- Rhagades : Presence of fissures around mouth.
- Saber shin : Presence of bowing of tibia.
- Clutton's joints : Bilateral knee joint effusions.

Diagnosis :

1. VDRL test : Compare VDRL titers of baby and mother.
   If there is 4 fold increase in baby → Congenital syphilis.
2. Rapid plasma reagent test (RPR).

Treatment :

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- IV Procaine penicillin (crystalline penicillin G) for 10 days.
- Ceftriaxone (if penicillin not available).

Active space

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Congenital Zika infection

0042:00

Transmitted by mosquito (Aedes aegypti mosquito).
Other modes : Sexual, blood borne, materno fetal transmission.
Risk of transmission high in 1st trimester.

Features :

1. Skull deformities (partially collapsed skull + overlapping sutures + prominent occiput).

2. Brain issues (neurotropic virus) :
   - Thinning/atrophy of cerebral cortex (especially white matter).
   - Hypertonia (spasticity).
   - Sensorineural hearing loss.
   - Extrapyramidal manifestations (chorea, athetosis).
   - Subcortical calcifications.

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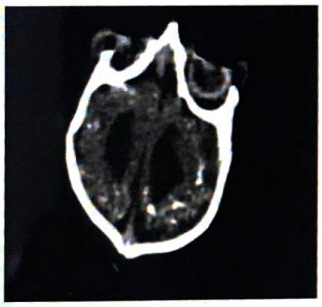
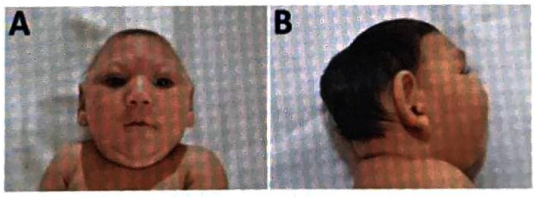
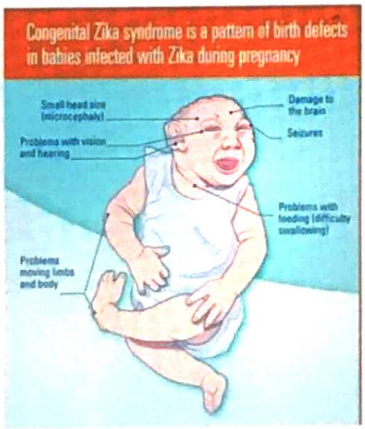
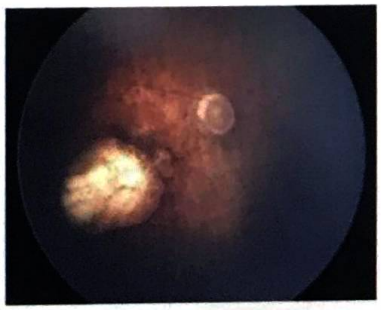


Skull deformities



3. Musculoskeletal findings : Contractures in lower limbs/ upper limbs → Deformed limbs.
   Lots of contractures → Arthrogryposis multiplex congenita.





Pigmentary retinal mottling

Active space

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4. Eyes :
   - Pigmentary retinal mottling.
   - Macular scarring.
   - Thinning of retina.
   - Coloboma/cataract.
   - Microphthalmia.

Diagnosis :
   - rRT PCR (r : Real time) – Investigation of choice.
   - Igm testing (if Igm positive, confirm with PRND (Plaque reduction neutralization testing).

No treatment.

No prophylaxis/vaccine.

Avoid exposure (prevent travel to endemic area).

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Active space

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COVID 19 IN CHILDREN

MOHFW (Ministry Of Health & Family Welfare) , Government Of India updated the guidelines in January ,2022.

COVID appropriate behaviour :

3Ws & 2Vs.

Watch your distance >2 metres.

Wear mask.

Wash your hands frequently.

Ventilation (open space ventilation decreases spread more in comparison to a poorly ventilated place).

Vaccination (Vaccination drive of 12-18 years of age has been started).

In India vaccination drive for 15-18 years old started on 3rd January 2022 and for 12-14 started on 16th March 2022.

COVID-19 symptoms in children 00:02:14

Features :

- Fever, cough, rhinorrhoea.
- Sore throat/irritation : Seen commonly with omicron variant.
- myalgia/headache /malaise.
- Diarrhoea.
- Loss of sense of smell/taste : Seen commonly with delta variant.

COVID-19 is mostly mild/asymptomatic in children.

moderate/severe disease is rare in children.

Active space

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Severity grading :

	Asymptomatic (incidental detection or H/o contact)	mild (URI)	moderate	Severe
Respiratory rate	Normal	Normal	Increased	Increased
SPO2 in room air	≥ 94%	≥ 94%	90-93%	< 90%
Danger signs/ symptoms	Absent	Absent	Absent	Usually present

Danger signs & symptoms : Grunting, lethargy, seizures, somnolence, severe chest retractions.

Asymptomatic patients are those children in which COVID-19 was an incidental finding or they have a history of contact with COVID-19 patient.

Mild COVID refers to upper respiratory tract symptoms.

Criteria for fast breathing :

Age	Respiratory rate
< 2 months	> 60/min
2-12 months	> 50/min
1-5 years	> 40/min
> 5 years	> 30/min

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Investigations :

CT chest is not indicated in paediatric covid.

Exception : If the child is terminally ill.

Severity of disease	Investigations
Asymptomatic	Not required
mild	Not required
moderate	CBC, ESR, chest X-ray, blood glucose.

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Severe	CBC, ESR, chest X-rays, blood glucose. CRP/ferritin (markers of inflammation), D-Dimer assays (for thrombosis), LFT/RFT (for multi-organ dysfunction).

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Management

00:09:55

	Asymptomatic	mild	moderate
Site	Home isolation	Home isolation or a covid care centre ± teleconsultation sos	Ward in a COVID 19 hospital or DCHC (Dedicated COVID Health Centre).
Treatment	No specific treatment	Symptomatic treatment (e.g. Paracetamol for fever or warm saline gargles for throat irritation)	O_a inhalation (goal of SpO_a : 94-96%). Fluid & electrolyte balance. Steroids only indicated if progressive disease course is noted. Antimicrobials if there is strong evidence of bacterial infection.

Severe COVID management :

Patients need to be admitted in ICU or High dependency unit (HDU) of COVID-19 hospital.

- O2 support (SpO2 : 94-96%).
- Fluid & electrolytes.
- Corticosteroids : Indicated in all severe cases.

Steroids to be used in Right time, Right dose, Right duration.

Active space

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Right time : Steroids are avoided in first 3 – 5 days after start of symptoms as they prolong the viral shedding.

Right dose :

Dexamethasone is recommended as 0.15 mg/kg to a max. dose of 6mg/kg once daily.

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Methyl prednisolone is recommended as 0.75 mg/kg to a max. dose of 30mg/kg once daily.

Duration : Steroids need to be given for 5 –7 days and then tapered over 10 –14 days.

- Anticoagulants are not usually indicated in management of all covid cases.

Only indicated in cases of thrombosis which are confirmed and D-dimer levels are elevated.

Low molecular weight heparin /LMWH (Enoxaparin) given in the dose of 1 mg/kg twice daily (BD).

- Antimicrobials are only indicated when there is a strong clinical evidence/ suspicion of bacterial infection.

Exception : If child has septic shock, antimicrobials can be used.

- Antivirals/monoclonal antibodies (Remdesivir, Favipiravir) are not recommended as their efficacy is not known in children.

Multisystem inflammatory syndrome in children (Mis-c)

00:20:25

Usually presents 2–4 weeks after acute COVID.

Risk factors :

- Age < 1 year.
- Comorbidities : Asthma , chronic lung disease.
- Obesity.
- Children with immunodeficiency.

Active space

Diagnostic criteria (WHO) for mis-c :

- Children and adolescents (0-18 years of age) with fever (temperature >38°C) for ≥ 3 days.
- Any 2 of the following should be present :
  1. Evidence of mucocutaneous inflammation like rash/ bilateral non purulent conjunctivitis.
  2. Hypotension or shock.
  3. Myocardial dysfunction, myocarditis, valvulitis, coronary involvement (e.g. aneurysms), elevated troponin.
  4. Evidence of coagulopathy (elevated D-dimers).
  5. Evidence of severe gastrointestinal involvement (uncontrolled abdominal pain /diarrhoea).
- Child should have additional findings :
  1) Increased inflammatory markers :
     a. CRP > 5 mg/dl.
     b. ESR > 40 mm/hr.
     c. Procalcitonin.
  2) Evidence of recent COVID-19 (RTPCR positive) or contact with confirmed COVID-19 case.
  3) No evidence of serious bacterial infection.

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Management of mis-c :

- Hospitalization is required in ICU.
- Methyl prednisolone (2mg/kg/day) is the steroid prescribed. Tapered over 2-3 weeks.
- IV Immunoglobulin (IVIG) is indiacted in all cases of mis-c Given at the dose of 2g/kg as slow IV infusion. Maximum dosage : 100g.

Active space

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CONGENITAL ANOMALIES AND HYDROCEPHALUS

Anomalies of nervous system :

Neural Tube Defects/NTD 00:00:15

most common congenital malformations in CNS.

Incidence : 1.5 per 1000 live births.

Increase risk in subsequent pregnancies : 3.5 to 4 per 1000 live births.

Neural tube development : Between day 24 to day 26 of gestation.

Neuroectoderm
↓
Neural plate
↓
Neural groove
↓
Neuropore
↓
Anterior neuropore (cranial structures like brain) | Posterior neuropore (caudal structures like spinal cord)
↓
Neuropore is closed
↓
Neural tube

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Basic defect : Failure of closure of neuropore.

Risk factors :

- Maternal deficiency of folic acid.
- Maternal usage of valproate and carbamazepine during pregnancy.
- Maternal intake of alcohol during pregnancy.
- Maternal overt and uncontrolled diabetes.
- Genetic defect : Mutation of the gene coding for methylene tetrahydrofolate reductase (MTHFR gene).

Active space

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Types of neural tube defects :

- Cranial/upper NTD
- Caudal NTD : more common.

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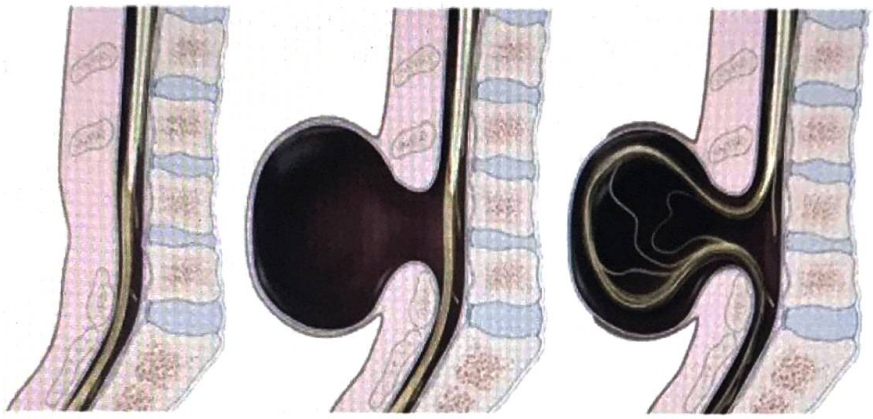
Caudal neural tube defects

00:06:16

Subdivided as :

Spina bifida occulta (hidden).

Spina bifida cystica (aperta) is of two types meningocele, meningomyelocele.



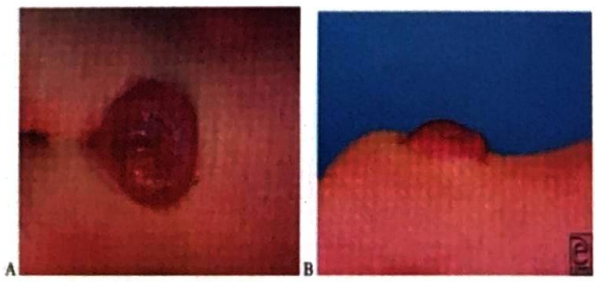
Spina bifida occulta meningocele myelomeningocele

Spina bifida occulta :

Some part of vertebral bone (posterior part) is missing.

It is hidden because the skin is covering the defect and is not visible externally.

Caused due to failure of formation of posterior vertebral arches.



Site of the defect : L5 or S1.

In children : Usually asymptomatic.

As spinal cord and meninges are well covered.

Clues for detection :

A tuft of hair over the defect.

Swellings like Lipoma or hemangioma over the defect.

Active space

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Spina bifida cystica/aperta :

Meningocele :
Defect in vertebral arches : Meninges protrude out through the defect and form a cyst.
More common in the lumbosacral region.
CSF filled lesion : Shows transillumination positivity.
Soft and fluctuant mass.

Meningomyelocele :
Spinal nerve roots also come out of the defect in the vertebral bone.
Usually associated with neurological deficits.
In lower limbs : Decrease in the tone of muscles.
In severe cases : Flaccidity of lower limbs.
Deep tendon reflexes are reduced or absent.
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Bowel and bladder incontinence.
Associated with Arnold Chiari type II malformation.
Seen in 80 % of cases.
Hence hydrocephalus is seen.

Cranial neural tube defects

00:13:48

Anencephaly :
Absence of brain/cerebral tissue.
Absent skull (calvarium).
The brain stem, cerebellum and spinal cord remain intact.

Active space

Subtypes :

1. Holoanencephaly :
   - Common.
   - Whole-brain is absent.
2. Partial anencephaly/meroanencephaly :
   - Part of the brain is present.

Encephalocele :

Part of the brain tissue comes out of the skull and forms a cavity.

Most common location : Occipital.

Iniencephaly :

Retroflexion of head : Due to defect in occipital bone and fusion of the cervical spinal vertebra.

Antenatal Ultrasound : Star gazer posture.

Cranial NTD : Invariably associated with severe neurological deficits.

Antenatal diagnosis :

USG :

By 14 to 16 weeks of gestation.

The earliest neural tube defect to be identified in antenatal USG : Anencephaly at 10 to 12 weeks of gestation.

Maternal serum markers :

Alpha-fetoprotein : Sensitive marker.

Acetylcholine esterase : Specific or best marker.

NTD prevention :

Folic acid supplementation.

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- Dose : 0.4 mg daily or 400 micrograms daily.
- If the mother has a previously affected child : 4000 micrograms daily or 4mg daily.
- Periconceptional supplementation : Should be started one month before planning of conception and continued till 12 weeks of gestation.

Active space

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Neuronal migration defects 00:21:13

Lissencephaly/Agyria :

Lis refers to smooth. Lissencephaly : Smooth brain (No sulci & gyri). Hence aka Agyria (absence of gyri).

Schizencephaly :

Schisis refers to split. Schizencephaly : Split-brain → Clefts in the cerebral hemisphere.

Porencephaly :

Presence of cystic spaces in the brain due to cerebral infarction in intrauterine life.

Agenesis of corpus callosum :

Corpus callosum is an interspheric connection. The defect is due to injury or insult to the commissural plate.

Features of agenesis of corpus callosum :

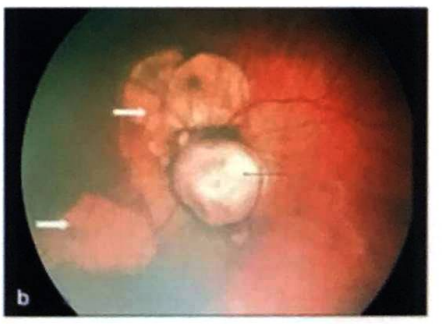
- Microcephaly.
- Low IQ.
- Increased risk of refractory seizures.

Imaging of the brain :

- Normal appearance of lateral ventricles : Bow tie appearance.
- In agenesis of Corpus callosum we see parallel lateral ventricles known as racing car appearance/ moose head appearance.

Aicardi syndrome 00:27:37

X linked, lethal in males.



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Activate Space

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Triad :

- Agenesis of corpus callosum.
- Chorioretinal lacunae :
  Punched out lesions seen in retina & choroid.
- Infantile spasm :
  EEG : Hemi hypsarrhythmia.

Other abnormalities :

- Coloboma of the optic disc.
- Retinal pits.
- Vertebral defects. E.g. Hemivertebrae.

Holoprosencephaly :

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Defect : Failure of the division of the forebrain.

Genetic defect : Sonic hedgehog gene.

Midline defect is seen on face :

- Fused eyes : Cyclops.
- Malformed nose : Proboscis.
- Midline cleft lip/palate.

Hydrocephalus 00:31:53

An increase in CSF volume results in an increase in the size of ventricles.

Normal CSF volume :

50 ml : Infants

150 ml : Adults.

CSF pathway :

CSF is formed by ultrafiltration of plasma.

Active space

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Secreted by choroid plexus of lateral ventricle → Foramen of monroe → Third ventricle → Aqueduct of sylvius → 4th ventricle → Foramen of Magendie → Central canal of spinal cord.

From 4th ventricle passes through foramen of Luschka → Subarachnoid space → Villi → Absorption.

Hydrocephalus causes :

Non obstructive causes/communicating hydrocephalus :
Increased production :

- Choroid plexus papilloma.

Reduced absorption :

- Blood in subarachnoid space (Subarachnoid hemorrhage), interferes with the absorption of CSF.
- Pus in subarachnoid space : In meningitis.

Obstructive/non communicating hydrocephalus (more common) :
Overall MC cause of Congenital hydrocephalus is Aqueductal stenosis.
Other anomalies :

- Dandy walker malformation.
- Arnold Chiari type II.
- Vein Of Galen (VOG) malformation.

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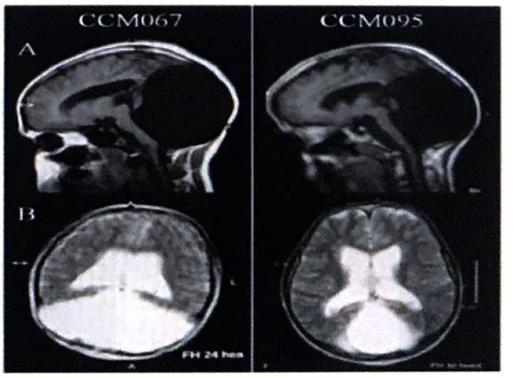
Dandy-Walker malformation

00:38:23



Defect in the development of the roof of the 4th ventricle.

Active space

This leads to expansion of the posterior fossa.

Features :

- The prominence of occiput is seen (bulging).
- Transillumination is positive.
- Cerebellar hypoplasia : Ataxia, dysarthria, nystagmus, vertigo.

Brain imaging :

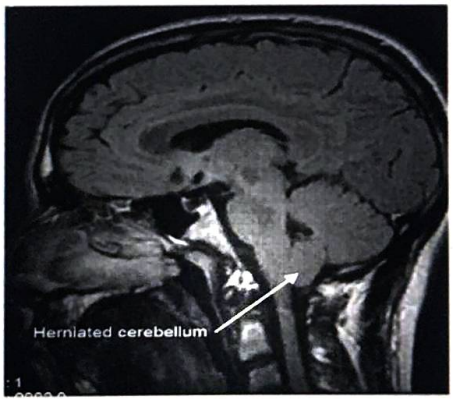
- The characteristic fluid-filled lesion in the posterior fossa of the brain.
- The size of the ventricles is increased.

Arnold Chiari malformation 00:41:22

Two types : Type I and type II.

Type I :

- Herniation of cerebellar tonsils through foramen magnum.
- This is not associated with hydrocephalus.
- Seen in adults.
- Presents with headache and neck pain.



Type II :

- Associated with hydrocephalus.
- Herniation of cerebellar vermis (hallmark for diagnosis is herniation of inferior cerebellar vermis), pons, medulla and 4th ventricle.
- Elongation/stretching of the 4th ventricle leads to its kinking or obstruction resulting in hydrocephalus.
- Associated with meningomyelocele : In 80% of cases.

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Active space

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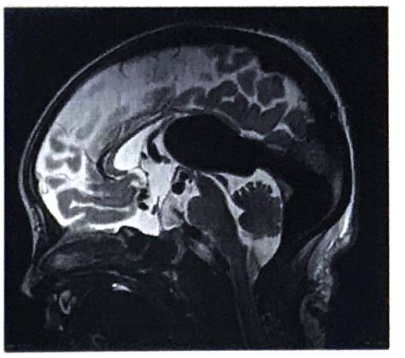
Vein of Galen malformation

00:44:58

Persistence of MPV : median prosencephalic vein (precursor of Vein Of Galen). This abnormal dilatation of VOG leads to obstruction of the CSF pathway resulting in hydrocephalus.



Increase in venous pressure in the brain : Cranial bruit, prominent carotid pulsations.
High pressure venous blood returns to right atrium resulting in cardiac failure.

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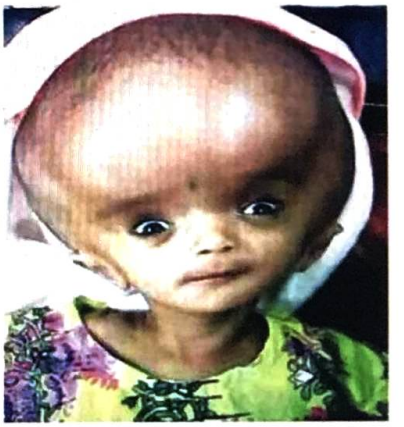
Classical clinical findings : Cranial bruit, prominent carotid pulsations.
IOC : MR angiogram (venogram) shows dilated VOG.

Clinical features of hydrocephalus

00:48:44

Macrocephaly : Large size of the head.
Head circumference (HC) > +2.5 standard deviation for age.
Abnormal increase in head circumference : HC increases by > 2cm/month.



Raised ICT :

- Dilated scalp veins.
- Bulging of anterior fontanelle.
- Wide separation of sutures.

Sunset sign : Eyeball is rotated downwards and the upper part of the sclera is visible. Dilated suprapineal recess

Active space

Leave Feedba

causes compression on the tectum of the midbrain.

Crackpot resonance/Mac Ewan's sign :

After 3 months of age. Gentle tapping on the skull results in a sound similar to the cracking of a pot.

Investigations :

Screening neurosonogram : USG is done through fontanelles.

Definitive/gold standard investigation : MRI brain.

Treatment of hydrocephalus 00:53:08

Medical :

Drugs to reduce CSF production :

Osmotic diuretic : Acetazolamide (carbonic anhydrase inhibitor).

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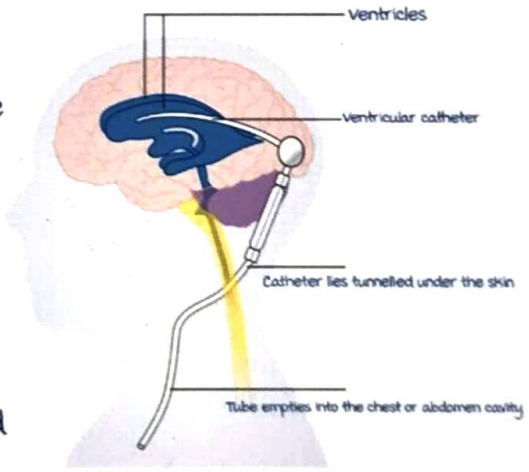
Surgical treatment : V-P shunting (Ventriculo-Peritoneal).

V-A shunting (Ventroculo-Arterial)

Complication of V-P shunting : Infection (shunt meningitis), caused by CONS (Coagulase Negative Staphylococcus). E.g., Staph. epidermidis.



Investigation to diagnose shunt meningitis :

Shunt tapping.

From ventricle, shunt is inserted & as it comes out, it is tunnelled subcutaneously. CSF is collected from the reservoir.

Preferred compared to lumbar puncture.

In aqueductal stenosis :

Endoscopic 3rd ventriculostomy can be done.

Here communication between 3rd ventricle & sub arachnoid space is made → CSF from 3rd ventricle shunted directly into sub arachnoid space.

Active space

Leave Feedba

SEIZURE DISORDERS AND EPILEPSY IN CHILDREN

Definitions

00:00:11

Seizure : Abnormal paroxysmal electrical discharge from neurons.

↓

Abnormal motor/sensory/behavioral/autonomic manifestations.

Detected by electro encephalogram (EEG).

Epilepsy :
Definition : Recurrent seizures (2 or more than 2 episodes 24 hours apart).
Most common cause in neonates : Hypoxic ischemic encephalopathy (HIE).
Most common cause in older children : Febrile seizures.
Most common type in neonate : Subtle (minimal features with eye blinking or deviation, chewing movements).
Most common type in older children : Generalized tonic clonic seizure (GTCS).

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Febrile seizures (FS) :
As per Association of child neurology (2021).
Definition :

- Condition with fever and seizure in age group 6 months to 6 years.
- Fever (temp > 38.4°C or 101°F).
- Without the evidence of CNS infection/electrolyte/ metabolic abnormalities.
- No h/o preceding trauma/afebrile seizures.

Active space

Types of febrile seizures :

1. Simple FS :
   - GTCS.
   - Lasting < 15 mins.
   - Does not recur within 24 hours.

2. Complex FS : Any of the following if present,
   - Focal seizure.
   - Lasts >15 mins.
   - Recurs within 24 hours.

Other definitions :

Febrile status epilepticus (FSE) : Seizure lasting for >30 mins.

FS plus : Seizure occurring after age of 6 years and/or afebrile seizures.

GEFS plus : Genetic or generalized epilepsy with febrile seizure.

Autosomal dominant inheritance : So family history of epilepsy with FS /FS plus.

Investigations for febrile seizure 00:09:23

In Simple FS investigations are not needed.

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The following investigations are indicated in :

- CBC : Complex FS or Febrile status epilepticus (FSE).
- Blood sugar or calcium estimation : CFS or FSE.
- Serum electrolytes (Serum Na^+) : FSE.
- Urine analysis : Only if <18 months without a focus of infection.
- MRI : Complex FS and Febrile SE.
  Done within 72 hours of onset of the seizure.
- EEG : CFS.
  Done within 1 week of onset of seizure.
- Lumbar puncture : Done when there is a suspicion of meningitis.

Active space

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1. Child <12 months of age :
   Who are unvaccinated for Streptococcus Pneumoniae and Hemophilus influenza type B.
   (H. influenza is a part of pentavalent immunization but not S. pneumoniae).
2. Child >12 months of age :
   With pretreatment with antibiotics.
3. Any age group :
   With suspicion of meningitis or in Febrile SE.

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Treatment of FS :

1. Control fever :
   - Antipyretics : Paracetamol : 10-15mg/kg/dose every 6th hourly.
   - Tepid sponging.
2. Control seizures :
   - If lasts for more than 5 minutes anti-epileptics are mandatory.
   - Intravenous Benzodiapines : Lorazepam / Midazolam (0.1 mg/kg).
   - Alternate routes :
     In hospital : Intranasal Midazolam.
     In community set up : Per Rectal Diazepam.

Prophylaxis (Prevention of recurrence) :

1. Intermittent prophylaxis :
   Oral Clobazam : 0.8 to 1 mg /kg/day for first 3 days of fever.
   Alternate : Oral Diazepam.
   Indications :
   1. Recurrent FS :
      3 or more episodes in 6 months or
      4 or more episodes in 1 year .
   2. Parental anxiety.
   3. Residence far from medical facility.

Active space

2. Continuous prophylaxis :

Daily usage of Sodium valproate.

Duration : 2 years of seizure free period before stopping medication.

Indications :

FSE

Frequent CFS.

FS with neurodevelopmental delay.

FS plus or GEFS.

Absence seizures 00:21:40

Also called 'Petit mal' seizure.

In age >5 years (usually onset 5-8 years).

Features at the time of seizures :

1. Vacant stares.
2. Unresponsive.
3. Eye flutter.
4. Lasts for few seconds (upto 30 seconds but clinically seen upto 10 seconds).

Precipitating factors :

1. Hyperventilation.
2. Stress/emotional problem.

Diagnosis :

- EEG : 3 Hz spike and wave pattern.
- Usually hyperventilation is done before EEG.

Treatment : Sodium valproate.

Alternate : Lamotrigine.

Atypical absence seizures :

- Myoclonic seizure component present.
- EEG characters : 1-2 Hz spike and wave pattern.
- Treated with Sodium Valproate.

Juvenile myoclonic epilepsy (JME) 00:25:23

Also called JANZ syndrome.

Typical age group affected : Adolescent.

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Age of onset : 12 to 18 years.

Mostly continues into adulthood.

Polygenic inheritance.

Active space

Leave Feedbac

Genetic association (Polygenic inheritance) :

- CLCN2 (related to chloride channels).
- CACNB4 (calcium channel/ calcium sensing receptors).
- GABRA1.
- myoclonin 1.

Features : 60c6b3eeaa8ded0e4e7e5ea7

Occurs upon awakening. .

Myoclonus : Sudden, jerky shock like contractions.

Normal consciousness (able to recollect events).

Can have other type of seizure patterns :

Generalized : GTCS.

Absence (Atypical) seizures.

Precipitating factors :

Sleep deprivation.

Alcohol consumption.

Diagnosis:

EEG : 4–6 Hz polyspike and wave pattern.

Treatment : Sodium Valproate.

Alternate : Lamotrigine / Levetiracetam.

Epilepsy syndromes 00:29:37

These are severe conditions with brain abnormalities predisposing to low IQ and developmental delay, posing difficulties in treating seizures.

1. West syndrome (Infantile spasms) :

Onset : Infancy.

Types :

Idiopathic/Cryptogenic :

Previously normal child .

Tend to have better outcome.

Symptomatic :

Associated brain disorder.

Worse outcome.

Active space

Features :

Spasm (sudden sustained contraction of muscles).

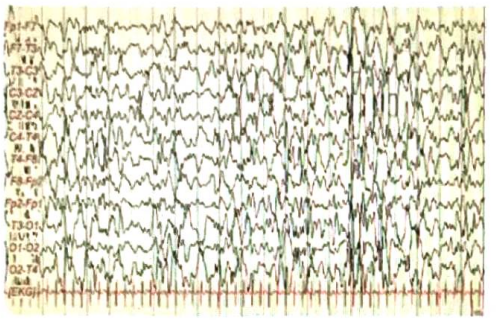
Jack knife seizures.

Repeated flexion of neck or Salaam spells.

Extension of neck (rare).

Triad :

1. Onset < 1 year of age characterized by infantile spasm.
2. Low IQ / developmental delay.
3. EEG : Hypsarrhythmia (high voltage multifocal spikes in a chaotic background).



Treatment :

- Injection ACTH (Ist line of drug).
- Vigabatrin.

If Infantile spasm is associated with Tuberous sclerosis (Ash leaf macule), then Vigabatrin is the drug of choice.

Dravet Syndrome :

Very severe form of epilepsy.

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Onset is in infancy in 1st year of age.

Also called severe myoclonic epilepsy in Infancy (SMEI).

Associated with genetic defect :

SCN IA defect : Alpha 1 subunit of sodium channel.

Clinical features : Typical pattern

Febrile seizures :

Starts at < 1 year of age.

Focal/ unilateral.

Clonic.

Active space

Leave Feedbac

Develops seizure with low grade fever (< 100°F).

↓

Later, afebrile seizures.

↓ 2nd year onwards

Multiple seizure patterns (myoclonic/absent type/any pattern).

Developmental delay.

Treatment : Difficult to treat.

Sodium valproate : Not satisfactory.

Stiripentol (latest) : Effective.

Lennox Gastaut syndrome :

Onset : 2 to 10 years of age .

Triad :

1. Developmental delay.
2. Multiple refractory seizures.
3. EEG : Typical 1.5–2.5 Hz spike and wave pattern.

Drug of choice : Sodium Valproate.

Status epilepticus

00:41:37

Definition: Prolonged duration >30 minutes.

Given in 2 time frames :

T1 : Lasting for 5 minutes or more.

Called as Impending SE.

Indicates the time after which the use of anti-epileptics is required to control seizure.

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T2 : Lasting for 30 mins or more .

Known as Established SE.

High risk of developing neurological sequelae.

Management :

In the first 5 minutes : No need of anti-epileptics.

Active space

Establish or secure :

- Airway.
- Breathing : (if SpO_2 <92%) : provide O_2.
- Circulation : Head turned to one side to avoid aspiration.
- Establish IV access.
- Assess hypoglycemia (blood sugar <54mg/dl).

↓

If hypoglycemia present , give 5ml/kg of 10% dextrose bolus.

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After 1st 5 minutes till 20 minutes :

IV Benzodiazepines are started.

Inj. Lorazepam /midazolam at 0.1 mg/kg/dose.

↓ Assess for 5 minutes , if not ceasing

Repeat dose.

↓ Seizure persisting (S++)

IV Phenytoin/

Fosphenytoin (preferred better because of lesser S/E)

at 20mg/kg as infusion over 20 mins.

↓ (S++) After 20 mins of onset.

IV sodium valproate / Levetiracetam at 20-40mg/kg.

↓ (S++) After 40 mins of onset .

IV phenobarbitone at 20mg/kg as IV infusion.

↓

Refractory Status epilepticus

Active space

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Refractory SE (seizure persisting after the above 3 medications).

A barbiturate coma is induced with the continous infusion of either :

1. Midazolam.
2. Thiopentone.
3. Phenobarbitone.

↓

If seizure still persist, it's a super refractory SE.

↓

Ketamine infusion.

or

IV immunoglobulin.

Active space

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183
Leave Feedba

CEREBRAL PALSY AND CNS INFECTIONS

Cerebral palsy

00:00:15

most common cause of motor disability in childhood.
It is a permanent, non-progressive damage due to a static (one-time) insult/injury to the developing brain resulting in abnormalities of tone, posture and movement.
It is an irreversible condition.

Cerebral palsy is also known as static encephalopathy.

Etiology :

Prenatal	Perinatal	Postnatal
CNS malformation	Birth asphyxia	CNS infections
Intrauterine infection (TORCH) affecting CNS.	Prematurity/low birth weight	Trauma
Teratogen (drugs) intake by mother	Birth trauma/ intracranial hemorrhage	Toxin mediated damage

more than 50% of the cases of cerebral palsy are due to prenatal etiology and less than 10% by perinatal etiology (previous highest)

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Clinical presentation :

m/c is Global developmental delay : Delay in at least 2 of the following milestones :

- Gross motor milestones.
- Fine motor milestones.
- Language milestones.
- Social milestones.

Active space

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Other findings :

- Persistence of neonatal reflexes (immature brain in CP → persisting immature/primitive/neonatal reflexes).
  Example : moro reflex persisting for more than 6 months of age (normal 5-6 months).
- Development of spasticity :
  Seen as clenched fist with cortical thumb (fist with thumb over palm). Cortical thumb, if noted after 4 months of age, considered to be abnormal.
- Scissoring of legs :
  Legs cross each other due to spasticity of the lower limbs.
- Abnormal posture due to spasticity.

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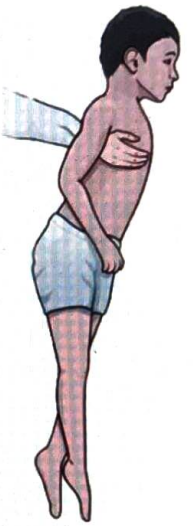
Cerebral palsy :
Scissoring gait



Co-morbid conditions related to cerebral palsy :

- Intellectual disability/Low IQ.
- Increased incidence of seizures.
- Microcephaly (poor brain growth).
- Abnormalities in speech/swallowing (can cause aspiration of feed = common cause of death).
- Increased risk of blindness/deafness.
- Increased risk of behavioral problems.

Classification of cerebral palsy :

- Spastic type :
  Most common type. Increased tone of the limb.

Active space

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Based on limbs affected :

1. Spastic quadriplegia.
2. Spastic diplegia.
3. Spastic hemiplegia.

- Dyskinetic type : Also known as extrapyramidal variety.
  Findings are similar to extrapyramidal damage such as :
  1. Dystonia/dyskinesia/rigidity.
  2. Chorea/athetosis.

  Caused by birth asphyxia affecting the basal ganglia.
  Intellect/IQ appears to be normal.
- Ataxic type :
  Due to cerebellar damage. Manifests with ataxia, slurring of speech, nystagmus and incoordination.
- Mixed type :
  Spastic + Extrapyramidal type presents together.

Spastic quadriplegia : Overall m/c type in India.

- All 4 limbs affected.
- Usually seen in term babies with H/O birth asphyxia.
- Severe type.
- High incidence of co-omorbidities (poor outcome).
- MRI findings :
  Immediate neonatal period : Parasagittal infarction seen.
  Older children : Multi cystic encephalomalacia.

Spastic diplegia :

- Both the lower limbs affected.

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- 2nd m/c type of cerebral palsy in Indian population.
- Seen in preterm babies with H/O birth asphyxia.
- During development, commando crawling is seen.
  Baby uses only the trunk and upper limb for crawling. Lower limbs are dragged with the body (mimics commandos while training).
- IQ of these children usually is normal and has good prognosis.
- MRI finding : Periventricular leukomalacia.
  Brain white matter surrounding the ventricles are affected.

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Spastic hemiplegia :

- One half of the body is affected (upper and lower limb on same side).
- Seen due to a vascular insult leading to perinatal stroke event.
- Early hand preference is seen (before 1 year).
  Since one side of the body is weak, the other side is always preferred (usually hand preference is attained at 3–4 years of age).
- MRI : Focal changes & porencephalic cysts due to infarcts.

Management of cerebral palsy :
Since it is an irreversible permanent damage, symptomatic management involving a pediatrician, physiotherapist, ENT surgeon, Ophthalmologist (multidisciplinary) can be given.
For spasticity : Drugs like Diazepam, Baclofen, Inj. Botulinum toxin A.
For contractures : Tendon release procedures.
Dystonia : Trihexyphenidyl, Levodopa.

CNS infections

00:19:27

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Bacterial meningitis :
Etiology based on age of the child :

- 0–2 months : Gram –ve bacteria like Klebsiella and E. coli.
- 2–24 months : Strep. Pneumoniae > H. Influenza.
- >2 years : Strep. Pneumoniae > N. Meningitis.

Overall, most common bacterial cause after neonatal period : Strep. pneumoniae.

In case of :

- Complement system defects (C5–C8) in a child : Meningococci is the most common cause.
- T-Lymphocytes defects (DiGeorge Syndrome or SCID or HIV) : Listeria Monocytogenes is the m/c cause.
- Child with splenic dysfunction due to chronic hemolytic anemia or asplenia) : High risk for encapsulated

Active space

bacterial infections with,

1. Strep. pneumoniae (most common).
2. Meningococci (N. meningitidis)
3. H. Influenza b.

- CSF leak (Rhinorrhea or otorrhea) :
  most common cause is Strep. pneumoniae.

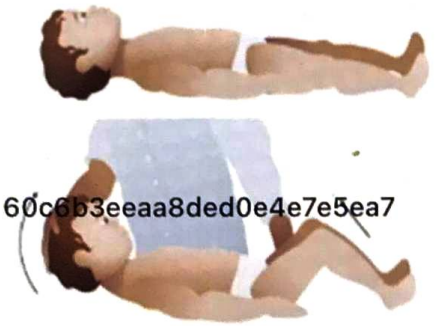
Features :

- Fever.
- Generalized seizures like GTCS.
- Irritability.
- Headache.
- Photophobia.
- Signs of raised intracranial tension (ICT) seen as bulging of anterior fontanelle.

Note : Suspect meningitis in a highly irritable child < 1 year of age, with fever & bulging anterior fontanelle.

- Meningeal irritation : Seen as
  1. Neck stiffness/neck rigidity.
  2. Brudzinski sign : Passive neck flexion → reflex flexion of hip & knee is seen.
  3. Kernig's sign : Flex hip & knee → passive knee extension (hip remains flexed) → resistance because of pain.

Signs of meningeal irritation are uncommon in children < 2 years of age.



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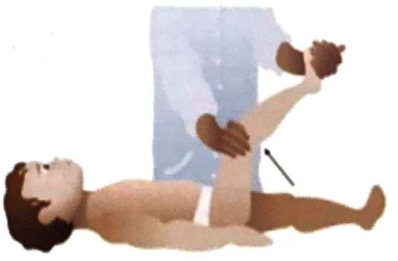
Brudzinski's sign:
Flexion of the hips and knees in response to neck flexion



Kernig's sign:
Resistance to extension of leg while the hip is flexed

Active space

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Investigations in a suspected child :

- Gold standard : CSF analysis by Lumbar Puncture (LP)
  In cases of ventriculoperitoneal shunt with suspeted shunt associated meningitis, shunt tapping is done.
  LP contraindicated → raised ICT patients → herniation and possibly death.
  Do CE-CT → shows meningeal enhancement.

CSF parameters	Normal	Bacterial meningitis	Reason
Pressure	<28 cm H_2O	Increased	
Appearance	Clear	Turbid	Presence of pus
Cells	< 5cells/cumm	upto 1000 cells/cumm	mostly neutrophils
Protein	20-45 mg/dl	upto 500 mg/dl	
Sugars		<40 mg/dl	Called hypoglycorrahia.

- Culture of CSF can be done to isolate the bacteria.
- PCR-CSF test to detect the bacterial antigen.
  Sensitive test & highly recommended.

Complications :

Most common acute complication : Seizures (seizures can be a complication & a feature of meningitis).

Other complications include :

- Subdural effusion/empyema.
- Hydrocephalus.
- Brain abscess (in immunodeficient children).
- Sensory Neural Hearing Loss (SNHL), especially noted with Strep. pneumoniae > H. influenza.

Treatment of bacterial meningitidis :

IV antibiotics (emperical treatment protocol) :

3rd gen cephalosporins (Cefotaxime/Ceftriaxone) → No response in 48-72 hrs → Add Vancomycin.

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Duration of treatment is around 10 days and in neonates for 3 weeks.

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Steroids :

Dexamethasone : 0.15 mg/kg/dose every 6 hrs for 2 days and usually started along with first dose of antibiotic.

Role : Decreases the inflammatory edema → decreases ICT.
Also decreases the incidence of complications, especially SNHL.

TB meningitis 00:34:26

most severe form of TB during childhood.

Pathophysiology :

Primary Infection
(Example : Pulmonary focus)
↓
Hematogenous dissemination
↓
Rich's focus in the brain
↓
Rupture of Rich foci
(during stress/low immunity like infection, severe malnutrition)
↓
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Seeding of meninges.

Thick exudates at the basal cisterns : Characteristic feature.

TB meningitis occurs in 3 phases :

1. Prodromal Phase :
   Lasts for 1–4 weeks, characterized by fever, irritability and vomiting.
2. Meningitis :
   The child will have high grade fever, signs of meningeal irritation, headache and photophobia.
3. Coma.

Note : Progression through each stage is not mandatory.
Higher the stage, poorer the prognosis.
If treatment is started in prodromal phase → Complete recovery is the rule.

Active space

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meningitis phase → 80% of children recover → 50% develops sequelae.

Stage of coma → Only 50% recover → 80% develops sequelae.

Neurological sequelae include obstructive hydrocephalus (Thick exudates → blocks CSF flow in subarachnoid area), cranial nerve palsies, optic atrophy, focal neurological deficits.

Prognosis depends on stage of detection.

Diagnosing TB meningitis :

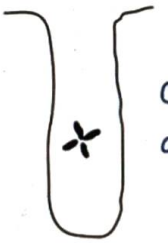
CSF analysis :

- ↑ CSF pressure.
- ↑ cells, up to 500/cu mm. mostly lymphocytes (after 48hrs). Neutrophils seen between 24-48 hours.
- ↑ protein, up to 3000 mg/dL.
- ↓ sugar.

Special tests :

- Chloride levels will be very low/undetectable.
- ↑ ADA (adenosine deaminase) levels.
- CSF culture and ZN staining (identify acid fast bacilli).
- Gene Xpert or CBNAAT test (Catridge Based Nucleic Acid Amplification Technique)

Cob web appearance of the CSF : Characteristic of TB meningitis.



Cob web appearance

CSF findings may mimic bacterial meningitis or may appear normal in 10-15% of cases each.

Perform neuroimaging → Basal exudates + hydrocephalus.

Treatment :

ATT for 10 months.

Steroids : Dexamethasone to decrease the incidence of neurological sequelae.

Active space

Viral infections of CNS

00:43:34

Virus usually causes meningoencephalitis (focal seizures or change in mental status indicative of brain involvement).

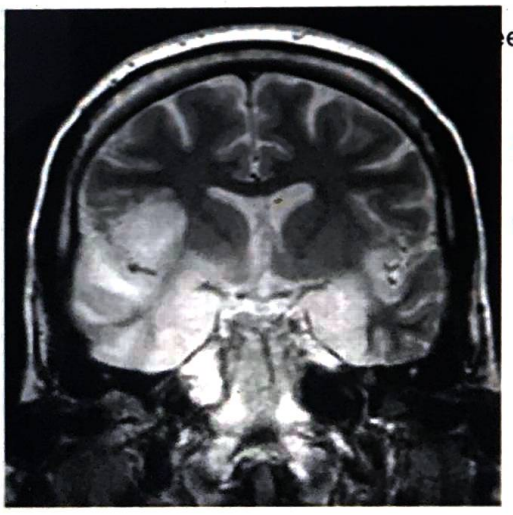
Overall, most common cause :
In India : Japanese encephalitis B (by culex mosquitoes).
Worldwide : Enterovirus.
Sporadic : HSV (1 >> 2).

CSF analysis :

- CSF usually appears to be clear.
- Pressure increased.
- Cells ↑ up to 100 cells/cumm and predominantly of lymphocytes.
- Protein levels are slightly increased up to 200 mg/dL.
- CSF sugar levels are usually normal.

PCR testing : Investigation of choice for HSV encephalitis.
JE virus specific IgM antibodies : Investigation of choice for Japanese encephalitis.

Herpes simplex encephalitis :
High propensity to involve the temporal lobe.
On EEG, characteristic finding PLED (Periodic Lateralizing Epileptiform Discharge) is noted.



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Herpes Simplex Encephalitis

IV Acyclovir (20 mg/Kg every 8 hrs x 14–21 days) is the drug of choice for HSV.

Active space

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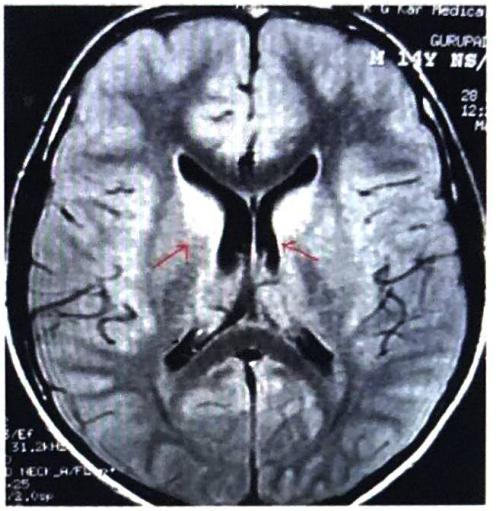
Japanese encephalitis :

Lesions are spotted surrounding the basal ganglia and thalamus.

The characteristic area affected in basal ganglia is the caudate nuclei. Treatment is only supportive.



Japanese encephalitis

Brain Abscess 00:49:50

Very severe disease.

Predisposing factors :

- Cyanotic congenital heart disease (Eg : TOF).
- middle ear infections (CSOM), mastoiditis.
- Sinusitis.
- Immunodeficiency.

Important associations :

Parietal lobe abscess : Seen in congenital cyanotic heart disease.

Temporal lobe abscess : Seen in CSOM, mastoiditis.

Organisms causing infections : Anaerobes >> aerobes.

Anaerobic : Anaerobic streptococci and anaerobic gram-negative organisms.

Aerobic : Aerobic streptococci, enterobacteriaceae, staphylococci.

Active space

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Brain abscess presents as a triad of,

1. Fever.
2. ↑ ICT.
3. Focal neurological deficits (abscess is localized to only one part of the brain).

Investigations :

Investigation of choice : MRI brain with contrast.

Abscess appears as a ring enhancing lesion with perilesional edema.

Large abscesses show evidence of midline shift.

Lumbar puncture is contraindicated due to high risk of herniation.

Treatment :

IV 3rd generation Cephalosporin (Cefotaxime/Ceftriaxone) along with Vancomycin and Metronidazole (for anaerobes).

Treated for a duration of 4-8 weeks.

If the child does not respond or presence of multiloculated abscesses → surgical drainage of abscess is done.

Surgical options are,

1. Burr hole surgery.
2. Craniotomy.
3. Stereotactic aspiration (newer modality).

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Active space

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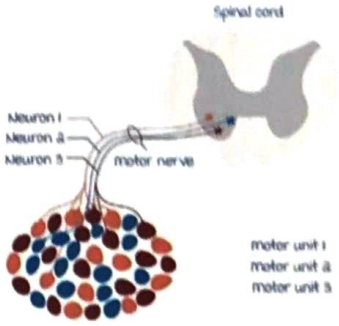
NEUROMUSCULAR DISORDERS

Introduction 00:00:09

Disorders of :

- Anterior horn cells called as neuronopathy (as it is composed of motor neurons).
  - Eg : SMA (Spinal Muscular Atrophy).
  - Poliomyelitis (Our country is declared free).
- Motor nerve called as neuropathy.
  Problem is in myelination of nerves
  - Eg : GBS (Guillain Barre syndrome)
- Neuromuscular junction.
  - Eg : Myasthenia gravis.
- In muscles called as muscular dystrophy.
  - Eg : Duchenne muscular dystrophy.



Spinal muscular atrophy (SMA) 00:02:32

Autosomal recessive inheritance.

Genetic defect related to chromosome 5q (long arm).

Survival motor neuron (SMN) gene is defective.

Function of SMN gene :

- Inhibits programmed apoptosis of motor neuron or prolong life of motor neurons.
- Gene is defective → Increased rate of apoptosis of motor neuron.

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Types of gene :

SMN 1 : Codes for functional protein.

SMN 2 : Codes for partly functional or dysfunctional proteins (non-functional protein).
Due to Defective exons.

Active space

Types of SMA : Based on onset of symptoms.

Type 0 :

Prenatal

Leads to foetal death → Severe SMA.

Intrauterine death.

Type 1 (Classical) :

0–6 months of age.

Werdnig Hoffman disease.

Death by 2–3 yrs of age.

Type 2 :

6–18 months of age.

Dubowitz disease.

Survival till school age.

Type 3 :

>18 months of age.

Kugelberg Welander disease.

Survival till teenage or early adulthood.

Severity decreases.

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Clinical features (Classical type)

00:07:38

Death of motor neuron → No impulses reach muscles → Flaccidity or decreased muscle tone.

Classical type :

- Decreased muscle tone.
  1. Normal child →
     - Active movement seen.
     - Resistance to passive movements.
  2. SMA child →
     - No active movement.
     - Frog leg position is seen.
     - No resistance to passive movement.
     - Ragged doll appearance :
       Hands, legs, head fall down on holding up the baby indicating no muscle tone.
- Decreased deep tendon reflex.
- Atrophy (disuse atrophy).

Active space

Leave Feedba

- Sparing of extraocular muscles.
- Fasciculations in tongue (mild tremor like).
- Brain unaffected, issues start at level of spinal cord, normal IQ/mentation.
- Feeding difficulty due to decreased muscle tone → Risk of aspiration → Death.

Treatment

00:12:14

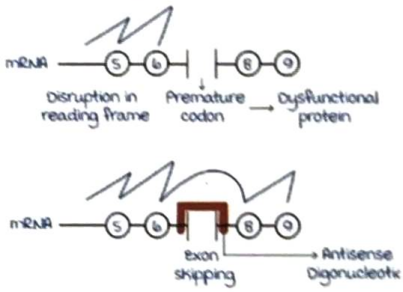
Not curable.

Symptomatic/supportive therapy usually.

Newer advances :

- Gene therapy :

  Normal SMN gene → Integrated into a vector 'Adenovirus' genome → Transferred to patient → Gene from adenoviral genome gets transferred to human genome → Normal copies of SMN gene synthesized.



- Exon skipping therapy :

  (Antisense oligonucleotide administration therapy)

  Eg : SMN 2 gene (dysfunctional protein coding gene)

  Defective exon 7 in mRNA → Exons are read from the frame → Disruption occurs due to missing exon 7.

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Disruptions → Premature stop codon → Dysfunctional protein.

In this treatment antisense oligonucleotide will occupy the area of defective exon.

The reading frame will not sense the defective exon 7 and gets disrupted instead will skip onto next exon (exon 8).

Active space

Protein formed is a partly functional protein (not 100% functional) and some amount of protein activity present.

↓

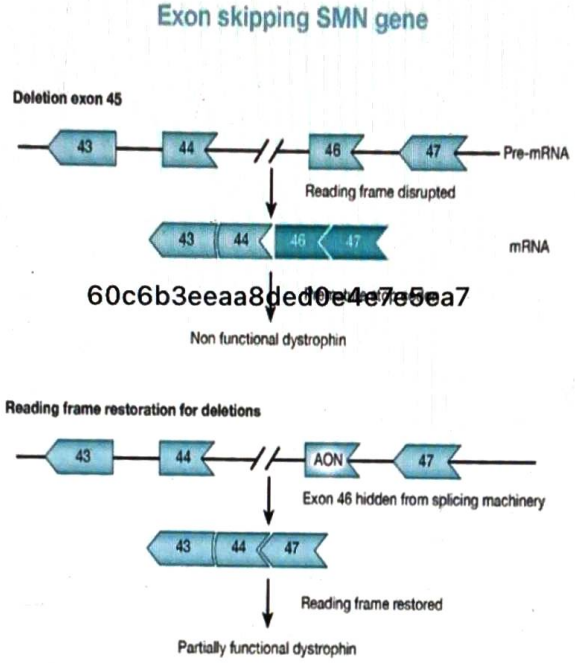
Helps in survival of motor neurons for a longer period of time.

↓

Translates to improvement in patient's weakness.

↓

Aids in child with SMA.



Antisense oligonucleotide used in SMA is Nusinersen.

Guillain Barre syndrome (GBS) 00:18:42

AKA Acute inflammatory demyelinating polyneuropathy (AIDP).

- Damage to myelin that is essential for nerve conduction.
- GBS occurs 5-14 days post-infection.
- Infections :

  GI → Campylobacter jejuni.

  Respiratory → Mycoplasma pneumoniae.

  Also considered to be a T cell mediated autoimmune disease
- Delayed hypersensitivity → T cell activation → B cell stimulation → Autoantibodies produced → Target again myelin → Demyelination.

Leave Feedba

- Associated antibodies :
  (Anti GM 1) & (Anti GD 1) → Antiganglioside antibodies.

- Features :
  Decrease in nerve conduction velocity so no proper muscle innervation.

  1. Acute Flaccid Paralysis (AFP) :
     i. Acute onset paralysis a/w decreased tone.
     ii. Ascending progression.
     iii. (LL → Trunk → UL → Face).
     iv. Symmetrical weakness.
     v. Typical pattern of weakness → Landry's paralysis.
     vi. Peaks by 2 weeks after onset.
     vii. Duration < 4 weeks (self-limiting).
     viii. Other cause for AFP is Poliomyelitis :
     Fever present.
     Asymmetrical weakness.

  2. Poly neuropathy → Nerves affected :
     i. Motor.
     ii. Sensory (Tingling , numbness, parasthesia, neuropathic pain).
     iii. Cranial nerves (B/L facial weakness.)
     iv. Autonomic (Changes in BP, HR).

- Unusual features : 60c6b3eeaa8ded0e4e7e5ea7
  1. Fever absent.
  2. Extraocular muscle (EOM) not involved.
  3. Bowel/bladder symptoms.

- Investigations :
  Electro physiological studies :
  Nerve conduction velocity → Decreased.
  Single fibre Electromyogram (EMG) → Evidence of denervation of muscle.

  CSF analysis :
  Not usually done.
  If done after 1 week of onset of disease,
  Albumino cytological dissociation seen.

Active space

Albumino cytological dissociation :

Discrepancy between cell count and protein.

Increased levels of CSF protein but as no infection cell count of CSF is normal/<10 cells per cubic mm.

Treatment :

Usually self-limiting within 4 weeks.

- IVIG (modality of choice) : To decrease progression of muscle weakness.
- Plasmapheresis : If not responding to IVIG .

Variants of GBS :

- Acute motor axonal neuropathy (AMAN) → Only motor nerves affected.
- Acute motor sensory axonal neuropathy (ASMAN).
- Miller Fischer → Characterised by AntiGQ 1b antibody. Triad of Ataxia , Arefelexia and Opthalmoplegia (EOM affected).

Neuromuscular junction disorders 00:32:32

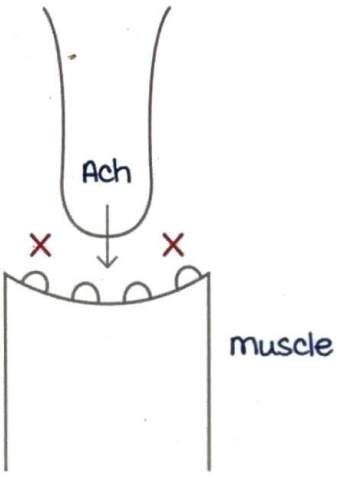
Synapse involved.

Juvenile myasthenia gravis (JMG) 60c6b3eeaa8ded0e4e7e5ea7

Features of myasthenia gravis in juvenile (< 18 years of age).

Pathophysiology :

- Autoimmune disorder.
- Usually, Ach released from nerve endings binds to the r ceptors & acts on the muscle.
- JMG → Anti acetylcholine receptor antibodies released → Occupy the receptor → Ach cannot act on muscle → Weakness.



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Characteristic features

- EOm involved → Earliest & constant.
  - Asymmetrical Ptosis (asymmetric weakness).
  - Ophthalmoplegia.
  - Diplopia.

- Weakness →
  - Diurnal variation (worse in evening).
  - Fatiguability :
    i. Exertion worsens weakness.
    ii. Worsens when child has illness or on medication like Aminoglycosides.
    iii. Skeletal muscles involving the limbs. (Proximal muscle groups)
    iv. Bulbar muscles → Aspiration.
    v. Respiratory muscles → Respiratory insufficiency.

Investigations :

- Anticholinesterase testing :
  - Administering of anticholinesterase improves weakness (Ptosis) immediately.
  - Anticholinesterase drug used → Edrophonium (tensilon)

Edrophonium :

i. Short-acting (response within 2 min).
ii. Not effective in < 1 yr of age as it is associated with adverse cardiovascular events.
iii. More time needed to record responses → Neostigmine used.

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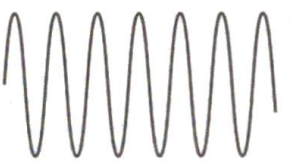
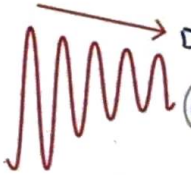
- Electrophysiological studies (EMG) :



Normal



Decremental Response.

MG

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Repetitive nerve stimulation → Low current given through nerve to innervate the muscle → Assess response → Performed repeatedly in a short span.

In JMG, a decremental response obtained (characteristic).

- Chest Imaging with Xray /CT :
  Associated with Thymic hyperplasia/Thymoma.
- Estimation of auto antibodies :
  Acetyl choline receptor antibodies common.
  Absent in some children, called Seronegative patients.
  In 40% of seronegative children, Anti MUSK is seen (muscle specific tyrosine kinase).

Treatment :

Will improve weakness but would not alter course of disease.

- Acetyl cholinesterase inhibitors (ACI) →
  Pyridostigmine.
  In newborns- Neostigmine(DOC).
- Immune modulatory medications → If no response to ACI.
  Steroids.
  Azathioprine.
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  IVIG.
  Plasmapheresis.
- Surgical → Thymectomy.
  Effective in seropositive children.
  Done within 2 years from onset of disease.

Other types (rare) :

- Congenital myasthenia syndrome :
  Inherited.
  Non-immune.
  Onset <2 years.
  Difficult to treat (no response to R_x).
  may worsen with acetylcholinesterase inhibitors.
- Congenital myasthenia gravis (transient) :
  Placental transfer of maternal antiacetylcholine receptor antibodies.

↓

Cross placenta.

Active space

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↓

Transient manifestation.

10 to 20% of mother with MG.

Starts within 3 days after birth.

Presentation :

Feeding difficulty.

Abnormal cry.

Bulbar/respiratory muscles weakness.

Complete recovery within 2 to 3 weeks.

Muscular dystrophy in children

00:49:21

Duchenne muscular dystrophy (DMD) :

- X-linked recessive (Female carriers, only males affected).
- Defect in short arm of chromosome Xp21.
- Dystrophin protein defective → Dystrophinopathies.
  Dystrophin → For coordination of muscular contraction.
- Based on severity of defect :
  Frameshift/nonsense (major) → DMD.
  Miss sense (minor) → Becker's muscular dystrophy.

Features :

- Onset < 5 years.
- Noticed when child starts walking or running around two years of age.
- Delay in attaining motor milestones (Walk, run).
- Characterised by proximal muscle weakness.
- Muscles affected → Thigh, calf (Waddling gait).
- Gower sign (non-specific) :

Child in lying or sitting position.

↓

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Asked to get up.

↓

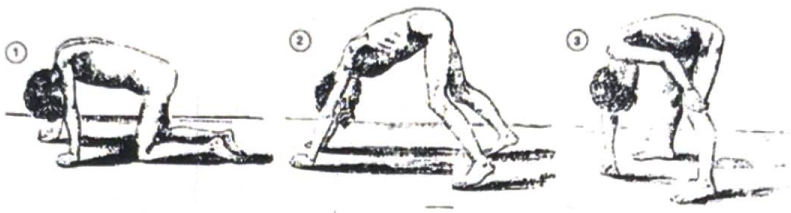
Uses upper limb over the thigh region as support due to proximal muscle weakness.

Active space

Conditions with positive gower sign :

- Duchenne Muscular Dystrophy.
- Juvenile Dermatomyocytosis .
- Steroid Toxicity.

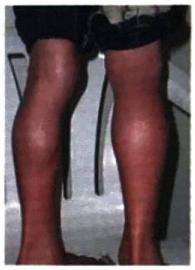
- Pseudohypertrophy → Prominent proximal calf area due to replacement of fibro fatty tissue on the apoptosed cal muscle.



- Scoliosis → When standing, trunk appears curved due to weakness of muscle.

Other findings :

- Slight decrease in IQ.
- Children are wheelchair-bound as they grow up.
- Develop complications like respiratory insufficiency+ Cardiomyopathy → Cardiac failure → Death.



Investigations :

- Release of muscle enzymes :

  Increase in Creatinine Phospho Kinase (CPK).

- Gene analysis.

Treatment :

Not curative but to decrease rate of apoptosis, steroids used

- Prednisolone.
- Deflazocort.

Exon skipping therapy (new treatment modality) →

Eteplirsen : Antisense oligonucleotide used in DMD.

	Duchenne muscular dystrophy	Becker muscular dystrophy
Mutation	Frameshift / nonsense	miss sense
Onset	< 5 years	> 5 years

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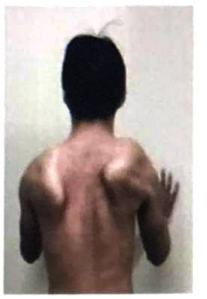
IQ	Decreased	Normal
Life Expectancy	Teenage	30–40 years

Becker muscular dystrophy is similar to DMD but a mild form.

Other muscular dystrophy (rare) 01:02:21

Facioscapulohumeral dystrophy :



Autosomal dominant inheritance.

Defective gene FSHD1.

Typical presentation :

- Facial weakness (earliest) :
  - Rounded mouth + Puckering (Protrusion of lips).
  - Unable to fully close eyes.
- Scapular weakness :
  - Asymmetrical winging of scapula.
- Upper limb :
  - Biceps, triceps (arm) weak, forearm spared.
  - Popeye arm appearance : Forearm appears normal but arm looks weak/smaller.

Association :

- Lordosis.
- Kyphoscoliosis.

These spine abnormalities are not specific for any muscular dystrophy.

Emery Dreifuss muscular dystrophy :

- X-linked recessive.
- Defective protein → Emerin, Laminin.
- Characterised by weakness :
  - Upper limb → Proximal muscles.
  - Lower limb → Distal muscles.

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Limb girdle muscular dystrophy :

- Autosomal recessive or autosomal dominant inheritance.
- Autosomal recessive →Children.
- Autosomal dominant → Adulthood.
- Defective protein → Calpain 3, Caveolin 3.

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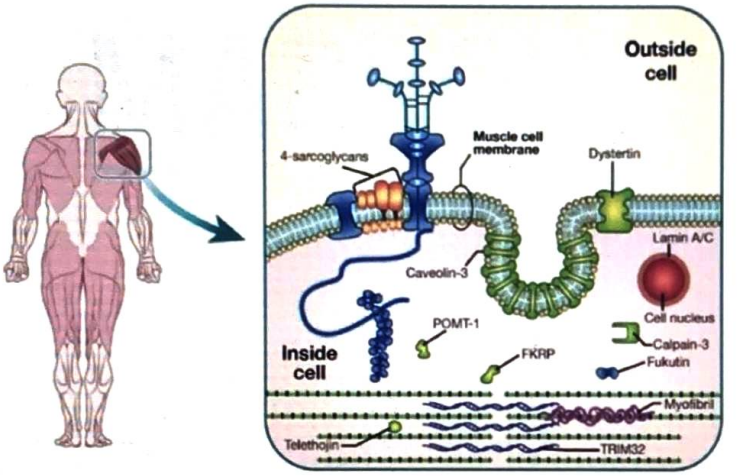
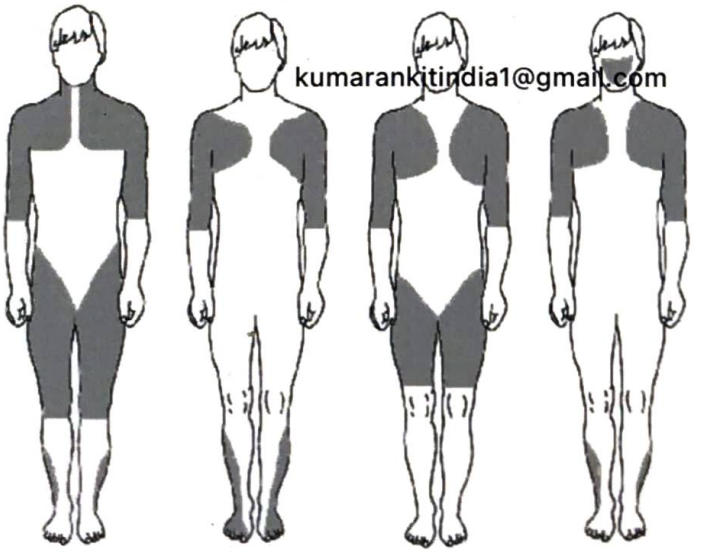
- Pattern of weakness :
  - Upper limb → Proximal muscles.
  - Lower limb → Proximal muscles.
- Involvement of pelvic girdle.
- Does not involve :
  - Pharyngeal/neck muscles (unlike in DMD).
  - Facial muscles.
  - EOM.
- Cardiac problems are rare (unlike in DMD).
- IQ/intellect → Normal (unlike in DMD).

Limb girdle muscular dystrophy



Single muscle cell



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Duchenne muscular dystrophy

Emery Dreifuss muscular dystrophy

Limb girdle muscular dystrophy

Facio scapulo humeral dystrophy

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GASTROINTESTINAL DISORDERS IN CHILDREN

GI anomalies 00:00:15

Esophageal Atresia :

Discontinuity in the esophagus because part of esophagus is underdeveloped/not formed.

Associations of Esophageal Atresia :

- TEF : Tracheoesophageal fistula.

Seen in 85% of cases of esophageal atresia.

- VACTERAL association :

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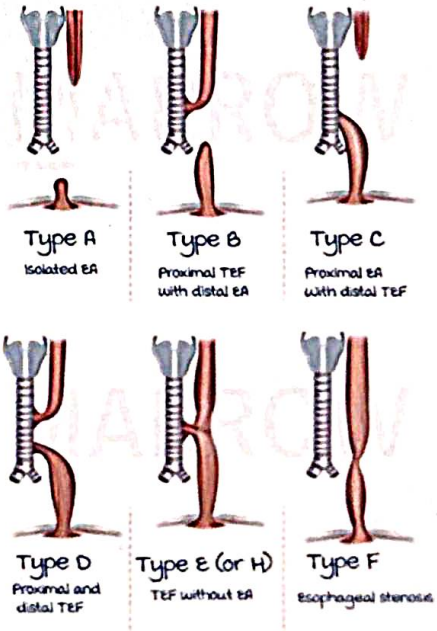
2. Anorectal malformation.
3. Cardiac defects.
4. Tracheoesophageal fistula (TEF).
5. Renal anomalies.
6. Limb defects.

Clinical presentation of esophageal atresia :

- Recurrent episodes of vomiting (Non bilious).
- Profuse drooling of frothy saliva in newborn.
- During antenatal period, mother has a history of polyhydramnios.

(During intrauterine life, the fetus is unable to swallow the amniotic fluid).

Complication : Repeated vomiting resulting in aspiration of feed, causing aspiration pneumonitis.



Classification of esophageal atresia with TEF :

Gross classification involves 5 types of tracheoesophageal fistula, out of which :

- Type C : most common type.

Active space

Proximal esophageal atresia with distal tracheoesophageal fistula.

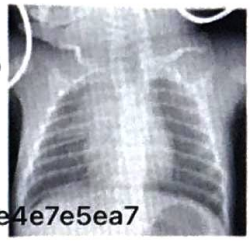
- Type D : Least common type.
- Type E : Also called as a H shaped TEF.

Diagnosis :

During routine checkup of the baby, while inserting a nasogastric tube, resistance is felt.

On X-ray, the classical coiling of the tube can be seen.

Diagnostic finding : Coiling of the NG tube in the upper esophagus.



Treatment :

Surgery is the definitive treatment : Posterolateral thoracotomy and repair of defects.

Idiopathic Hypertrophic Pyloric Stenosis 00:07:52

Hypertrophy of circular muscle fibers in the pyloric region, causing narrowing at the pylorus.

- more common in males than females (4:1 incidence).
- Risk of IHPS in 1st born male child is higher.
- Usage of macrolides (erythromycin) either by mother during pregnancy period or newborn baby (1st 2 weeks) is associated with IHPS.

Features :

- Non bilious forceful projectile vomiting after feeds.
- Vomiting never starts immediately after birth, it appears 2-3 weeks after birth.
- Epigastric olive shaped swelling is usually noted after the vomiting episode.
- Visible gastric peristalsis, due to increased force exerted by the smooth muscles to overcome the obstruction, usually noted after feeding. Usual direction of peristalsis is from left to right side.

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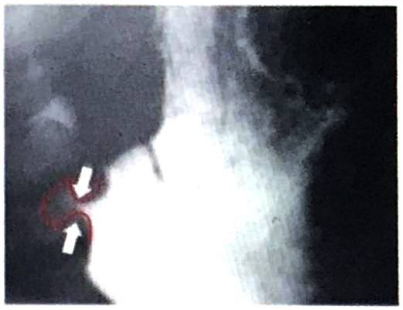
Association :

Jaundice : Seen due to decreased activity of the enzyme UDP-GT, resulting in increased unconjugated bilirubin and jaundice.

Icteropyloric Syndrome : Jaundice + IHPS.



Investigations for IHPS :

Contrast X-Ray :

1. String sign : Narrow pyloric channel is seen on contrast x-ray.
2. Double Track sign.
3. Shoulder sign : The hypertrophic muscle bulges into the gastric antrum.
4. Mushrooming : Bulging of the hypertrophic muscle into the 1st part of the duodenum.

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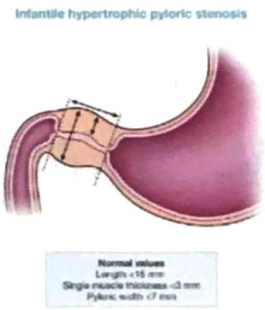
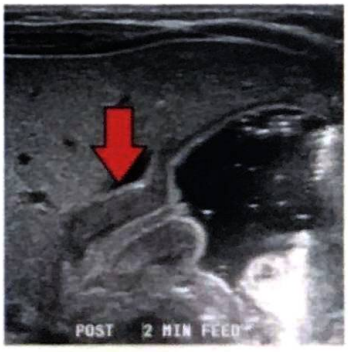
Investigation of choice : USG.

1. Thickness of the pyloric muscle should be ≥ 4 mm.
2. Length of the pyloric channel should be ≥ 16 mm.

Treatment :

- Manage dehydration and electrolyte imbalances before surgery for IHPS.
- In this patient hypochloremia with metabolic alkalosis and hypokalemia is seen as HCL is vomitted out.
- Presence of hyponatremia in this patient initiates conservation of sodium by reabsorption. Kidney excretes H^+ and K^+, therefore paradoxical aciduria is seen. (Hypokalemic hyperchloremic metabolic alkalosis with paradoxical aciduria).
- Dehydration : 1/2 NS + K^+ or Ringer Lactate used.
- Surgical treatment : Ramstedt's Pyloromyotomy.





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Duodenal Atresia

00:22:37

Due to failure of recanalization of duodenal lumen during fetal development.
Genetic association for duodenal atresia is seen in Down's Syndrome.

Down's Syndrome :
Most common anomaly : endocardial cushion defect.
2nd most common anomaly : GI anomaly, duodenal atresia.
30% of downs syndrome patients are associated with duodenal atresia.

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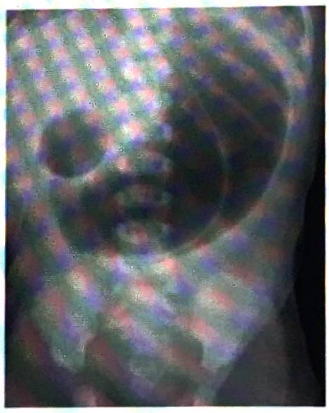
Other associations of duodenal atresia :

- Prematurity (25-30% of cases).
- Congenital heart defects are seen in 30% of cases.

Features of duodenal atresia:

- Bilious vomiting seen soon after birth.
- Not associated with abdominal distention.
- maternal history will be significant for polyhydramnios.

Investigation :
X-Ray :
Double bubble sign.
(This sign is also seen in cases of annular pancreas and duodenal web).
Diagnostic modality :
USG/CT imaging.



Treatment :
Surgery : Duodeno-duodenostomy.

Meckel's diverticulum

00:28:05

most common GI anomaly, seen in 2% of the population. It is a remnant of omphalomesenteric duct or Vitello intestinal duct.

This is a true diverticulum (includes all layers of the GI tract).

Features :

- Present in 2% of population.

Active space

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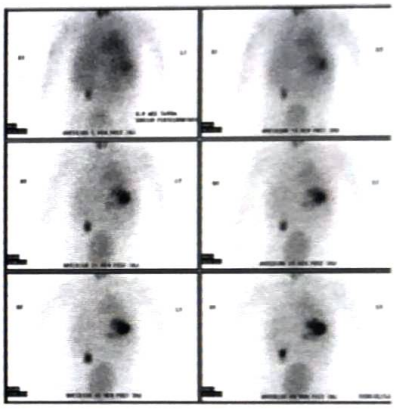
- Length of meckel's diverticulum is 2 inches (5 cm).
- It is 2 feet from the ileocecal junction at the anti mesenteric border of the ileum.
- Lined by 2 types ectopic/heterotopic mucosa :
    1. Gastric mucosa.
    2. Pancreatic mucosa.
- 2 times more common in females than in males.
- most of the cases are asymptomatic.
- If the child has symptoms, they usually appear before 2 years of age and seen as painless rectal bleeds or bleeding at the time of stool passage.
    1. Bleeding is due to deep ileal ulcers caused by hydrochloric acid secretion by ectopic gastric mucosa.
    2. Bleeding described as currant jelly stools or melanotic bleeds or maroon bleeds.
- Other rare forms of presentation include :

1. Intestinal obstruction due to meckel's diverticulum acting as a lead point for intussusception (ileocolic intussusception).
2. Volvulus :
Due to twisting occurring by the fibrous band connecting the meckel's diverticulum and the umbilicus.
3. Littre's Hernia :
When meckel's appears as a part of the hernia sac..



Intestinal obstruction due to intussusception is the most common presentation in adults.

Diagnostic investigation for meckel's :

meckel's scan - Technetium-99m (99mTc) pertechnetate scintigraphy.

Intestinal obstruction due to intussusception is the most common presentation in adults.

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Active space

Diagnostic investigation for meckel's :

Meckel's scan : Technetium-99m (99mTc) pertechnetate scintigraphy.

This radionucleotide has high affinity for gastric mucosa, therefore stomach and the ectopic meckel's gastric mucosa shows increased uptake.

Treatment :
Asymptomatic meckel's is discovered as incidental finding.
Treatment is done if :
1. If the meckel's diverticulum has a narrow mouth or a narrow base, then a diverticulectomy is done.
2. Patient is symptomatic.

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Hirschsprung disease

00:38:38

Also known as aganglionosis (absence of ganglions in the intestinal submucosal and muscular layers meissner's , Auerbach's plexus).
Occurs due to failure of migration of neurons during development and is known as neurocristopathy.

Due to absence of the ganglions, there is failure of relaxation of intestine. Therefore, the stool is unable to pass through the aganglionic segment of the intestine. Therefore, the stools are stuck to the intestine proximal to the affected area.

- more common in males (4:1; males: females).
- most commonly affects rectosigmoid area (80% cases).

Named as short segment disease, because only a short segment is affected.

- 15% cases, appear as long segment disease, where areas proximal to sigmoid gets affected (E.g. transverse colon).
- 5% of cases entire colon gets affected (total colon disease).

Associations :

- Down's syndrome.
- Mutations in RET proto oncogene.

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Clinical Features :

Rectosigmoid area is always in a constricted state.

In newborns :

1. This manifests as delay in the passage of meconium, even after 48 hours of birth.
2. Abdominal distention.
3. Rectum feels empty because of lack of stools.
4. Withdrawal of finger results in sudden expulsion of meconium.
5. In older children constipation is seen.

Investigation :

Rectal manometry : Absence of recto anal inhibitory reflex.

Best investigation for diagnosis : Rectal suction biopsy.

Findings :

1. Hypertrophy of nerve fibers.
2. Absence of ganglion.
3. Increased staining/activity of Acetyl cholinesterase.

Treatment :

Previously resection and anastomosis was done.

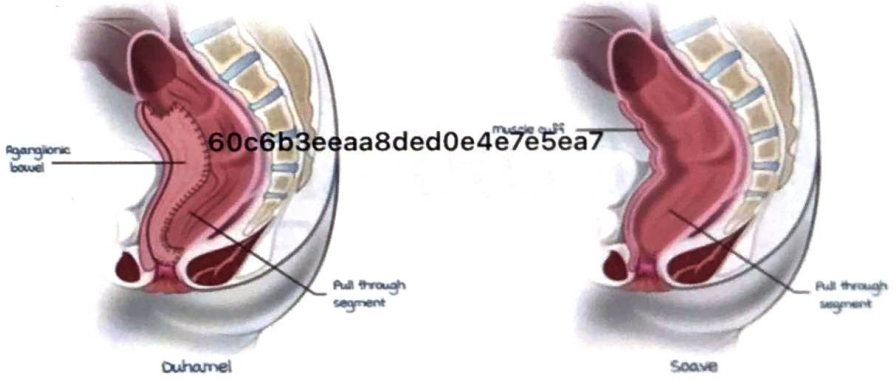
Nowadays pull through procedure is done :

Normal part of intestine is pulled through the aganglionic part and attached to the last part of intestine i.e. anus.

1. Duhamel procedure.

2. Soave procedure (preferred procedure).

In this procedure, 1st strip the rectal mucosa; leaving behind the muscular part of rectum (muscular cuff). This muscular cuff provides a support to the proximal part of intestine that would be joined to the distal part.



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DIARRHOEA

One of the important causes of under 5 mortality.

Common definitions

00:00:31

	Duration	Etiology
Acute diarrhea	< 7 days	Infection
Persistent diarrhea	> 14 days	Infection
Chronic diarrhea (rare)	> 14 days	Non infectious (often associated with malabsorption)

Acute diarrhea :

It is defined as loose stools/change in consistency of stools/or a child who used to pass normal consistency stool now passes watery stool.

Increase in frequency of stools > 3/day.

Commonest infections that cause diarrhea are viral >> bacterial.

Overall most common cause is Rotavirus.

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Other viruses associated with diarrhea in children include :

Norwalk virus, Adenovirus, Astrovirus, Calicivirus.

Overall most common bacterial cause in children is E.coli (ETEC).

Other bacterial causes associated with diarrhea in children include : Salmonella, Shigella, Campylobacter.

Parasite induced diarrhea are predisposed in children with immunodeficiency.

Most common parasite that cause diarrhea is Giardia.

Other parasites causing diarrhea are :

Entamoeba, Cryptosporidium (most common parasitic diarrhea in HIV positive children).

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Dysentery :

It is the presence of loose stools along with blood in stools.

Almost always bacterial in origin.

most common bacteria associated with dysentery in children : Shigella flexneri.

A child with diarrhea, most common cause is rotavirus, so no need to start antibiotics.

However, if the child has dysentery, it is almost always due to bacteria and should start antibiotics.

Dehydration

00:07:39

First step in management of diarrhea is to assess the child for dehydration.

Based on presence or absence of dehydration & severity of dehydration there are 3 types :

	No dehydration	Some dehydration (losing 50-100 ml/kg fluid loss)	Severe dehydration (fluid loss of >100 ml/kg)
	Active, alert	Thirsty, slightly irritable	Lethargic
Skin pinch test (skin turgor over abdomen)	Fast /< 1 sec	Slow /<2 sec	very slow/>2 sec

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Plan of management	Plan A : Replacement of ongoing loss by ORS. <6 months : 50 ml of ORS or ¼ of a glass for every loose stool. 6 months to 2 years : 50–100 ml of ORS or ¼ to ½ of a glass for every loose stool. >2 years : 200 ml of ORS or 1 glass for every loose stool.	Plan B : 1. Rehydration : ORS : At 75 ml/kg over 4 hours. 2. Replacement of ongoing loss by ORS similar to plan A. 3. maintenance/ daily fluid requirements Calculated by Holiday Segar formula (24hrs) : 0–10 kg : 100ml/kg 10–20 kg : 1000 ml + 50 ml/kg for every kg above 10 kg. >20 kg : 1500 ml + 20 ml/kg for every kg above 20 kg.	Plan C

Eg for Plan B : For a child of 22 kg,

fluid required = 1500 + 40 = 1540.

Plan C :

1. Rehydration is with IV fluids (since child is lethargic) : RL 20 ml/kg,
   100 ml/kg of fluid is given (30 ml/kg + 70 ml/kg).

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	30ml/kg	70ml/kg	Total
< 1 year	1 hour	5 hours	6 hours
> 1 year	½ hour	2 ½ hours	3 hours

2. Replacement of ongoing loss.
3. Daily fluid maintenance.

Active space

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Fluids are the cornerstone of treatment in diarrhea.
Fluid can be given orally as ORS or as IV fluids if child is not able to take orally.
In addition to fluids, zinc is recommended in the management of diarrhea.
Zinc helps in epithelialization of the GIT, it also helps decrease severity of loose stools as well as decreases the future recurrence of diarrhea in a child.
Dose :
< 6 months : 10 mg/day.
> 6 months : 20 mg/day.
Duration : 14 days irrespective of the age.

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No role of antimotility agents like Loperamide.
No recommendations for antisecretory agents like Racecodotril.
No guidelines or recommendation for probiotics.

ORS

00:19:22

WHO recommended low osmolarity ORS is being used now.
Total osmolarity = 245 mOsm/L.
ORS is composed of electrolytes and glucose, the composition of which is as follows :

Sodium = 75
Glucose = 75
Potassium = 20
Chloride = 65
Citrate = 10

Both glucose and sodium are kept in same concentration as in intestine the SGLT1/sodium glucose co transporter, transports 1 molecule of sodium and 1 molecule of glucose. Therefore, both are kept at 1 : 1 ratio.

Active space

Indications for anti microbial treatment

00:23:06

Dysentery : 3rd generation cephalosporin like Cefixime.
Other medications that can be useful are Ciprofloxacin and Azithromycin.

Severe malnutrition : Ampicillin + Gentamycin.

Cholera : Doxycycline.

Giardiasis/amoebiasis : Metronidazole.

Persistent diarrhea in children 00:26:12

It starts as an acute diarrhea but it persists for > 14 days and is also due to an infection.

It is predisposed in children with the following risk factors :

- Age < 1 year (6 months – 1 year) :
  This may be due to immaturity of GIT as well as immune system to overcome the infection which is why the infection persists for 14 days.

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- Low birth weight/LBW or Small for Gestational Age/SGA babies or malnutrition :
  Delayed mucosal healing of GIT this results in unusually prolonged diarrhea/persistent diarrhea.
- Some organisms :
  E.coli : Enteroadherent E.coli
  Enteroaggregatory E.coli.
  Salmonella.
  Shigella.

Lactose intolerance : It is both a risk factor as well as a consequence of persistent diarrhea.

CMPA/Cow milk Protein Allergy : β lactoglobulins, casein.

Consequences :

It can predispose to nutritional deficiency especially micronutrients like Vitamin A.

Imperative to give 1 dose of vitamin A while treating persistent diarrhea.

Lactose intolerance :

Acquired/secondary due to persistent diarrhea.

Lactose enters the GIT from milk consumed.

Lactose is digested with the help of enzyme, lactase.

Lactase is produced from the tip of gastrointestinal villi.

In persistent diarrhea there is loss of tip of villi leading to loss of lactase activity.

There is no digestion of lactose.

Active space

Leave Feedbac

The lactose is now made to pass down the GIT in its undigested form.

Once it reaches the colon, the colonic bacteria converts them into lactic acid and hydrogen.

Lactic acid can be excreted in the stools causing perianal excoriations/rash.

Hydrogen being gaseous substance, when excreted in stool causes explosive stools/gaseous stools.

Management :

ORS after assessing dehydration.

Zinc (same dose and duration as in acute diarrhea).

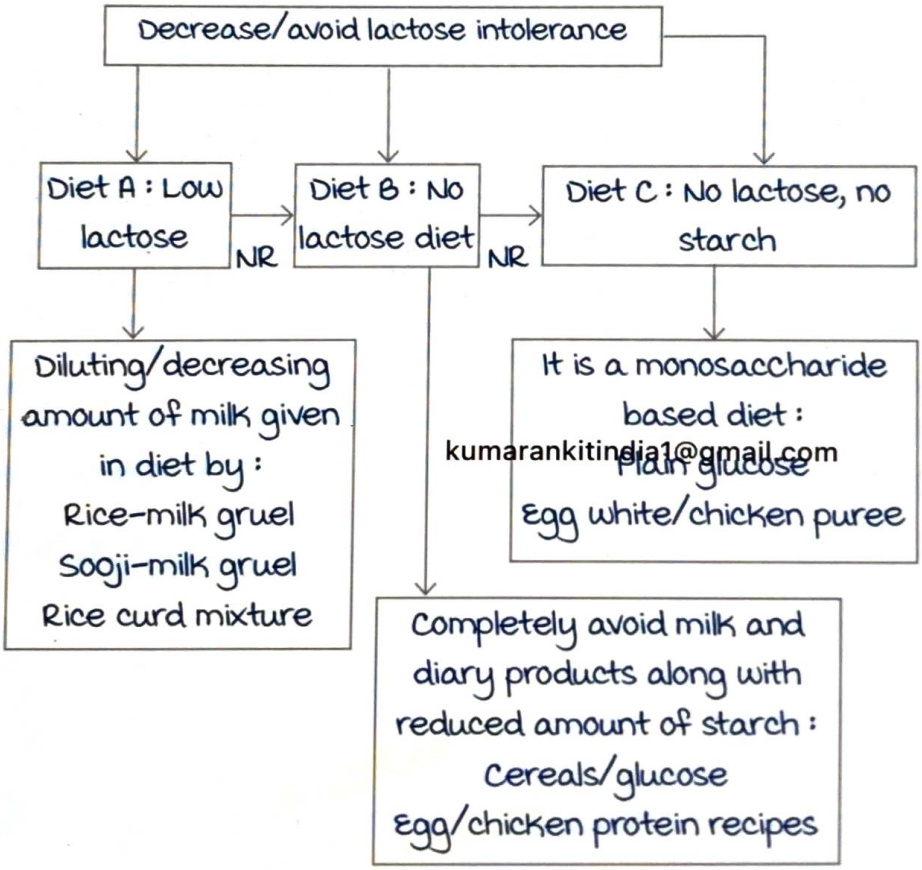
Vitamin A supplementation (single dose) :

< 6 months : 50,000 IU.

6 – 12 months : 1 lakh IU.

> 12 months : 2 lakh IU.

Decrease or completely avoid lactose in the diet. Done in 3 steps :



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NR : Not Responding

Active space

MISCELLANEOUS TOPICS IN GIT

Gastro Esophageal Reflux Disease 00:01:05

Reflux of the gastric contents to esophagus.

Child presents with recurrent vomiting.

Recurrent vomiting causes malnutrition and child presents with failure to thrive.

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Recurrent vomiting can also lead to aspiration pneumonitis.

Age at onset : < 6months (4 months).

Sandifer syndrome :

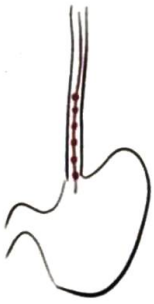
Associated syndrome with GERD.

Characteristically presents with sudden episodes of spasm of muscles that involved trunk and limbs and Opisthotonus posturing (arching of the back) seen similarly in tetanus, BIND.

Diagnosis :

1. 24 hour esophageal pH monitoring if ≤ 4
2. Best diagnostic test : Multichannel intraluminal impedance monitoring.

Pediatric multichannel intraluminal impedance monitoring of pH at different levels



Management :

Usually resolves spontaneously at 2-3 years.

Therefore, first choice include Non pharmacological measures :

- Thickening of feed.
- Small frequent feeds.
- Positioning
  1. If baby is awake, prone position is recommended.
  2. If baby is sleeping, supine posture with slight head end elevation (30 degrees).

Active space

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Pharmacological measures :
Tried only if non pharmacological measures fail.
Drug of choice : Proton pump inhibitors.
If Refractory GERD, does not responding to treatment :
Surgical correction in the form of fundoplication is done.

Foreign bodies in GIT

00:10:10

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Once in the stomach, 95% of all ingested objects pass without difficulty throughout the remainder of the GIT and excreted in feces.
Hence it is important to recognize any foreign body that is above the stomach i.e. In esophagus.
Age : 6 months – 6 years (2 years).
Most common objects : Coins and small toys.
Most dangerous : Disc shaped button batteries, they can cause caustic damage and coagulative necrosis of the esophagus.

Diagnosis :
History
Investigations

- X-ray : 90% of foreign bodies are radio opaque.
- Remaining 10% : Barium study or esophagogram.
- Radiolucent foreign bodies appear as a filling defect on contrast X-ray.
- Sites of impaction :
  1. At the level of cricopharynx.
  2. At mid-esophagus (at level of tracheal bifurcation).
  3. Just above the lower esophageal sphincter.

Presenting features :

- Dysphagia
- Odynophagia
- Drooling of saliva
- Chest pain.

Management :
If object is blunt and the child is asymptomatic : Observe for 24hours.

Active space

Indications for immediate removal :

1. Impacted for >24hr.
2. Sharp objects.
3. Button batteries.
4. Magnets.
5. Symptomatic at any time.

Removal is done by flexible endoscopic removal.

Foreign body in stomach and beyond :
mostly conservative management as they pass out through the intestine in the next 4-6 days.

Indications of endoscopic removal :

1. Battery >20mm diameter, Age <5 years : Cannot pass through the pylorus.
2. Double sided sharps objects like fish bones, toothpicks.
3. Open safety pin.
4. Size of the object, longer than >5cm : Cannot pass through the intestinal flexures.
5. Multiple magnets.

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Celiac disease

00:22:07

Gluten hypersensitivity (A kind of food allergy)
Pathogenesis : T-cell mediated autoimmune disorder, autoantibody mediated small intestinal destruction.
Foods with gluten (BROW) :

1. Barley.
2. Rye.
3. Oats.
4. Wheat.

Genetic predisposition : HLA DQ2 and HLA DQ8.
In these patients, gluten exposure can lead to celiac disease (especially seen in Northern parts of India where wheat is the major diet).
Features / Clinical profile :

1. Malabsorption leads to Failure to thrive.
2. Chronic diarrhea (>2 weeks).

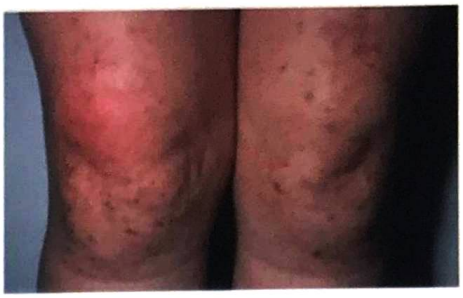
Active space

3. Anemia which is refractory to iron treatment. Iron, Vit B12, folic acid absorbed in small intestine.

Age at onset : >6months after birth, with commencement of complementary feed as breast milk doesnt have gluten.

Associations :

1. Hashimoto's thyroiditis.
2. Type I Diabetes mellitus.
3. Down's syndrome.
4. Turner's Syndrome.
5. Dermatitis Herpetiformis :



Papulo vesicular eruption on extensor aspects of joints.

Complications :

Increased risk of tumors in adult life :

- Adenocarcinoma.
- Non-Hodgkin lymphoma : T cell type.

Diagnosis :

Revised European Society of Pediatric Gastroenterology and Nutrition (ESPGAN)

1. Compatible Clinical profile

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2. Jejunal biopsy (investigation of choice in Indian setup).
   - Villous atrophy/ flat villi
   - Crypt hyperplasia
   - Increased intraepithelial lymphocytes
3. Antibodies (They are all IgA antibodies)
   - Anti-tissue transglutaminase antibody (tTg) screening test.
   - Anti-Endomysial antibody : Best or diagnostic.
   - Anti-Gliadin antibody. } High false positive hence
   - Anti-Reticulin antibody. } Little role in testing for CD
4. Unequivocal response in 12 weeks to gluten free diet.

Management :

Lifelong gluten free diet.

Follow up to check for compliance, post 6 months : Fall in titres levels of tTg antibody.

Repeat jejunal biopsy for follow up is not recommended.

Active space

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NEONATAL CHOLESTASIS

Neonatal cholestasis 00:00:15

Cholestasis : Stasis of bilirubin.

There is elevation of conjugated bilirubin levels manifesting as jaundice.

Definition : If the total serum bilirubin (TSB) is :

- < 5mg/dL, then increase in conjugated bilirubin to ≥ 1 mg/dL.
- >5mg/dL then increase in conjugated bilirubin to ≥ 20%.

It is suspected in a baby when the jaundice is persisting for > 14 days.

- Color of stools : Pale/clay/white stools (core of the stools is assessed because outer aspect of the stools can appear yellowish due to bile staining of the epithelial cells of the intestine).
- Color of urine : Dark yellow color.
- Associated with hepatomegaly.

Etiology :

Extrahepatic causes/obstructive jaundice: Due to obstruction to the flow of bile.

- Biliary atresia (EHBA, Extrahepatic biliary atresia) : Overall most common cause.
- Choledochal cyst.

Intrahepatic causes : Characterized by impaired excretion of bilirubin into the bile canaliculi.

- Idiopathic neonatal hepatitis (most common intrahepatic cause).
- Infections
  1. TORCH infections.
  2. Neonatal sepsis.
  3. UTI.
- Metabolic causes :
  1. Galactosemia.

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2. Tyrosinemia.
3. α 1 Antitrypsin deficiency (mc inherited cause worldwide).
4. Cystic fibrosis.
5. PFIC (Progressive Familial Intrahepatic cholestasis) : A genetic disorder characterized by impaired bile salt secretion. Subtypes :
   Type 1 : ATP8 B1 gene defect (Byler disease).
   Type 2 : ABC B11 gene defect.
   Type 3 : Adult onset.
   Both these subtypes have low levels of GGT (Gamma Glutamyl Transpeptidase) enzyme.

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- Genetic conditions :

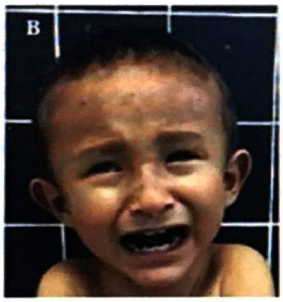
1. Down's syndrome/ Trisomy 21.
2. Edward syndrome/ Trisomy 18.
3. Alagille syndrome.

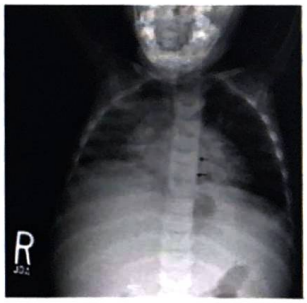
Alagille syndrome

00:13:12

Is an autosomal dominant inherited disorder.

Gene defects : JAGGED 1 (mc gene defect) /NOTCH2 defects.





Pathology : Paucity of intrahepatic bile ducts, probably due to immune mediated destruction of the bile ducts.

The number of bile ducts keep decreasing with age.

AKA Arteriohepatic dysplasia.

Physical examination findings :

- Jaundice.
- Abnormal facies : Broad forehead, deep set widely spaced eyes, long straight nose, underdeveloped mandible(triangular facies).
- Vertebral anomalies : Butterfly shaped vertebrae, hemivertebra.

Active space

- Eye : Microcornea, Posterior embryotoxon (it is seen as a ring shaped structure due to prominence of the Schwalbe's ring).
- CVS anomalies : Peripheral pulmonary stenosis (MC cardiac finding), Tetralogy of Fallot (TOF), Ventricular Septal Defect (VSD).
- Pruritis, tendon xanthomas (due to high cholesterol).

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Extra-hepatic biliary atresia 00:20:10

Biliary atresia not associated with cyst formation.
AKA Non cystic Obliterative Cholangiopathy. (Cystic Obliterative cholangiopathy seen in choledochal cyst).
MC variety : Complete obliteration of the extrahepatic bile ducts along with obliteration of intrahepatic ducts at the level of hilum.
Classification of biliary atresia (based on the presence of other associated anomalies) :

- Perinatal type (70% cases) : Not associated with any other anomalies.
- BASM syndrome (15% cases) : Associated with; (Poor prognosis).
  1. Biliary Atresia.
  2. Splenic malformations : Polysplenia.
  3. Situs inversus.
  4. Cardiac defects.
  5. Malrotation of the GIT.
- Other 15% cases : Associated with Renal anomalies and choledochal cyst.

Investigations 00:26:32

1. Liver function tests : Increased conjugated bilirubin.
2. Markers of cholestasis :

- 5' nucleotidase.
- Alkaline phosphatase (ALP).
- GGT : Normal is up to 40 U/L, amount of raise in GGT can give the clues to the cause of neonatal cholestasis.
  4 times elevation : Intrahepatic causes.

Active space

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10 times elevation : Extrahepatic causes.

Exception : GGT levels are low in PFIC type 1 and 2.

3. Ultrasound : High frequency USG (HUS) is more helpful in the evaluation of neonatal cholestasis.

USG is also helpful in detecting the associated anomalies.

Characteristic USG findings in EHBA :

- Triangular cord sign : Hyperechoic areas corresponding the fibrous/atretic portion of the bile duct.
  Non visualized gallbladder/ micro gallbladder/ gallbladder ghost.

Triangular cord sign

4. EHBA findings in Radionucleotide study :

HIDA scan (Hepatobiliary scan with Tc labelled imino Di-acetic Acid scan) shows decreased excretion of the tracer from the liver due to obstruction.

Phenobarbitone loading/priming has to be done for 5 days to improve the uptake of radionucleotide tracer by the liver. It is used only as a screening test and not a specific investigation.

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5. Best investigation : Liver biopsy.

	Intrahepatic/Idiopathic neonatal cholestasis	EHBA
Hepatic architecture (lobules)	Disarray of hepatic lobules	Normal
Giant cells	++++	+
Portal reaction	Inflammation with minimal fibrosis	Marked fibrosis & some lymphocytic infiltration
Neoductular proliferation	Rare	Marked
Others	Evidence of steatosis	Portal bile duct plugging along with bile lake formation.

Active space

Leave Feedback

Neonatal cholestasis : Management

00:37:53

medical management :

Clinical impairment	management
malabsorption of dietary long chain triglycerides	medium chain triglyceride (MCT) oil supplementation
Fat soluble vitamin malabsorption	Vitamin A, D, E, K supplementation. • VitaminA : 10,000–25,000 IU/day. • Vitamin D : 800 IU/day. • Vitamin E : 25 to 200 units/kg/day. • Vitamin K : 2 to 5mg repeated every 4 weeks.
micronutrient deficiency	Ca^{2+} phosphate and Zinc supplementation
Deficiency of water soluble vitamins	Twice the RDA.
Retention of biliary constituents such as cholesterol (pruritis)	UDCA (Ursodeoxycholic acid). Cholestyramine.

Surgical management :

Algorithm followed in surgical management of EHBA :

Exploratory Laparotomy

↓

Peroperative Cholangiogram

↓

Temporary procedure : Kasai Procedure/Hepaticojejunostomy

↓

Definite management : Liver Transplantation

Kasai procedure :

It is a temporary procedure.

Communication is created between the liver and the small intestine (jejunum) for the bile to drain out.

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AKA hepaticojejunostomy.

It should be done within 8 weeks after birth.

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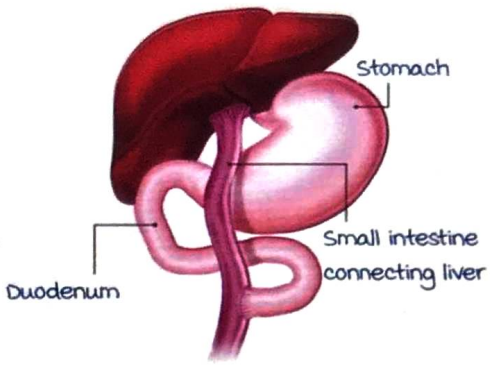
MC indication for Liver transplantation in children : EHBA.



Kasai Procedure

Active space

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METABOLIC LIVER DISORDERS

Alpha-1 antitrypsin deficiency 00:00:31

It is a commonly inherited metabolic liver disorder.
Less common in India compared to the West.
Inheritance : Autosomal recessive.
Gene defect : SERPINA 1 gene on chromosome 14q.
Production of a misfolded form of alpha 1 antitrypsin (α1 AT).

Normal role of alpha 1 antitrypsin :

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Alpha 1 antitrypsin (liver)
↓ Blood
Lungs
↓
Protects the lung tissue (acinus) from neutrophil elastases

In α_1 AT deficiency :

misfolded α_1 AT

- Accumulates in the liver → Toxic damage to the liver cells
- Cannot enter the lungs → Destruction by neutrophil elastases → Panacinar emphysema

Phenotypic expression of α_1 AT :
Normal phenotype : Pi MM (Pi = protease inhibitor, MM = normal allele).
Abnormal phenotype : Pi ZZ (ZZ = abnormal allele).

Features :

- Liver damage : In 80–85 % cases, the liver recovers completely (only 15–20 % cases develop chronic liver disease).

Active space

Leave Feedbac

- Pan-acinar emphysema : Does not develop in childhood. It develops in the adults by 30-40 years of age. Smoking increases the risk of development of pan acinar emphysema.

Investigations :

- α_1 AT levels : Low (< 50-80 mg/dL).

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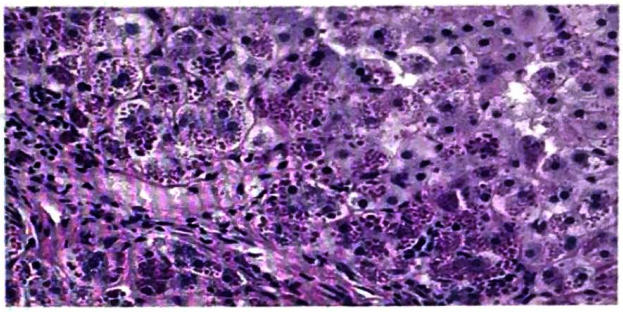
- Genetic testing : SERPINA 1 gene.
- Liver biopsy : Misfolded α_1 AT as intrahepatic globular inclusions in the liver on PAS stain (Periodic Acid Schiff positive). Diastase resistance is also seen.



Treatment :

Supportive treatment.

Limiting the exposure to tobacco smoke.

In adults : Infusion therapy with normal α_1 AT collected from healthy individuals.

If chronic liver disease develops : Liver transplant.

Wilson's disease

00:10:17

Gene defect : ATP7B on chromosome 13.

This gene codes for P-type adenosine triphosphatase.

Normal role :

- Excretion of the excess Cu^{2+} into the bile.
- Incorporation of Cu^{2+} with ceruloplasmin (Cu^{2+} binding protein). Bound Cu^{2+} in the blood is nontoxic.

Wilson's disease :

- Impaired excretion of Cu^{2+} in the bile leading to accumulation of Cu^{2+} in the liver (liver damage).
- Increased levels of unbound/free Cu^{2+} in the blood (toxic).

Active space

Clinical features :

- Chronic liver disease, cirrhosis of liver : Age at presentation is usually > 1-2 years.
- Eye damage : Kayser- Fleischer (KF) ring in the cornea due to deposition of Cu^{a+} in the descemet's membrane.
- Cataract : Sunflower cataract.
- Neurological manifestations :
  1. Basal ganglia damage : Abnormal movements, tremors, seizures.
  2. Psychiatric manifestations.

1st feature of brain damage noted in children with Wilson's disease : Deterioration of scholastic performance.
Childhood onset Wilson's disease : Classically presents with chronic liver manifestations.
Adult onset Wilson's disease : Presents with neurological manifestations.

Other manifestations :

- Cu^{a+} mediated hemolysis of the RBCs (negative Coomb's test).
- Arthralgia.

Investigations :
Screening investigations :

- Urine Cu^{a+} : Increased.
- Serum ceruloplasmin : Decreased.

Confirmatory investigations :
Liver biopsy :

- Macrovesicular steatosis.
- Mallory-Hyaline bodies (not very specific).

MC condition associated with the presence of Mallory body in liver biopsy in children : Wilson's disease.
Assessment of Cu^{a+} levels in the liver tissue : Increased (> 250mg/g dry weight of the liver tissue).

Treatment : Decrease the Cu^{a+} levels in the body by limiting the amount of Cu^{a+} in the diet.

Active space

Leave Feedba

Cu^{2+} chelating agents :

- D-Penicillamine : DOC in children.
- Triantene.

Zinc (induces production of metallothianine which inhibits the absorption of Cu^{2+} from the intestine).

CLD/cirrhosis : Liver transplantation.

Chronic usage of D-penicillamine leads to the deficiency of vitamin B6.

So, D-penicillamine should always be prescribed along with vitamin B6 to prevent the development of optic neuritis.

Glycogen storage disorders/GSD 00:23:19

Types : Liver GSD > Muscle GSD

Liver GSD	Muscle GSD
GSD 1 : Von Gierke's disease (Glucose-6 phosphatase deficiency). Overall MC type.	GSD 2 : Pompe's disease (Acid maltase deficiency).
GSD 3 : Cori's/Forbe's disease (debranching enzyme).	GSD 5 : McArdle's disease (muscle Phosphorylase) MC muscle GSD.
GSD 4 : Anderson's disease (branching enzyme).	GSD 7 : Tauri's disease (Phosphofructiokinase).
GSD 6 : Hers disease (liver Phosphorylase).	

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Common features of liver GSD and muscle GSD :

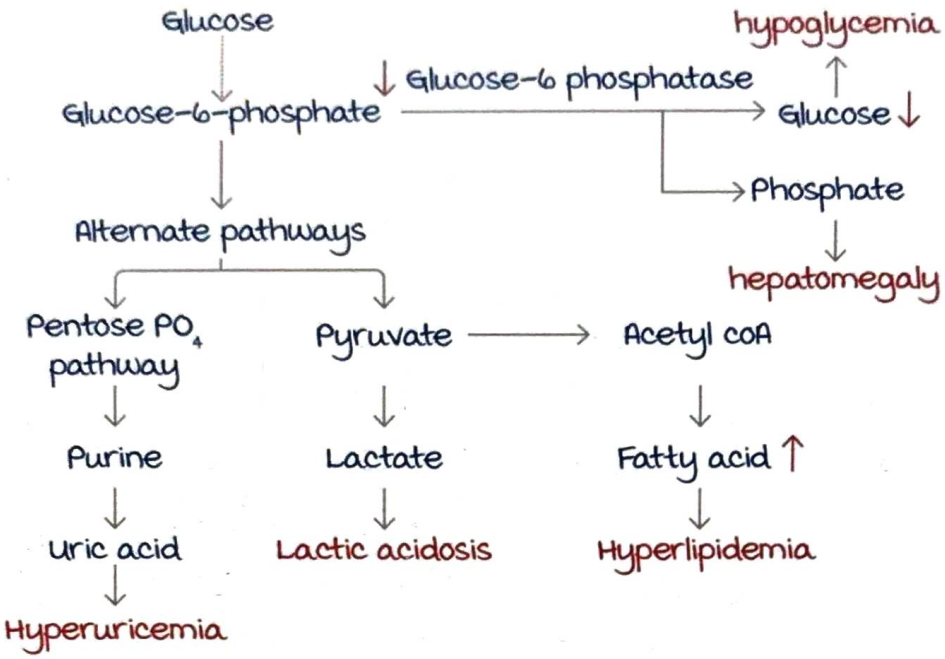
Liver GSD	Muscle GSD
Fasting hypoglycemia (hypoglycemic seizures).	Exertional fatigue.
Hepatomegaly.	Recovery of fatigue with rest (2^{nd} wind phenomenon).

Active space

GSD I/Von-Gierke's disease :

Deficiency : Glucose-6 phosphatase enzyme.

Overall most common type.



Features :

- Characteristic round facies (doll-like facies).
- Abdominal distension (hepatomegaly).

Biochemical findings :

- Hyperuricemia.
- Lactic acidosis.
- Hyperlipidemia.

USG :

- Hepatomegaly.
- Renomegaly.

Treatment :

- Frequent feeding (every 2-3 hours) to prevent episodes of fasting hypoglycemia.
- Night time : Nasogastric drip feeds (for infants).
- Uncooked corn starch in every meal (because it releases small amounts of glucose at frequent intervals).
- Older children : Waxy maize heat modified starch to prevent overnight episodes of hypoglycemia.

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Active space

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GSD 3 : Cori's/Forbes disease :
Deficiency : Debranching enzyme.
Features :
Some cases can also have muscle involvement.
No biochemical abnormalities.
No renomegaly.
Spleenomegaly is present.

GSD 4 : Anderson's disease :
Deficiency : Branching enzyme.
Liver damage and early onset cirrhosis.
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Worst prognosis.

GSD 2 : Pompe's disease :
Deficiency : Acid maltase (lysosomal enzyme).
Also a lysosomal storage disorder.
Skeletal muscles and cardiac muscles are affected.
Hypertrophic cardiomyopathy leading to cardiac failure.
Liver is also involved (hypoglycemia).
Macroglossia.
Severe hypotonia (floppy infant).
Poor prognosis.
Treatment : ERT (Enzyme replacement therapy).

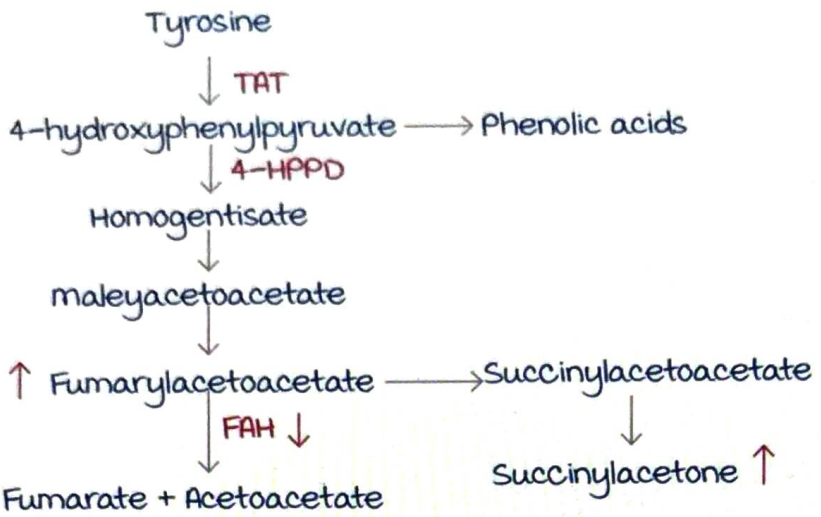
Tyrosinemia

00:45:04

Defect in tyrosine metabolic pathway.
3 Types : Type I is more common.
Type I tyrosinemia :
Autosomal recessive.
Deficiency of Fumaryl Acetoacetate Hydrolase (FAH).

Active space

metabolic pathway of tyrosine :

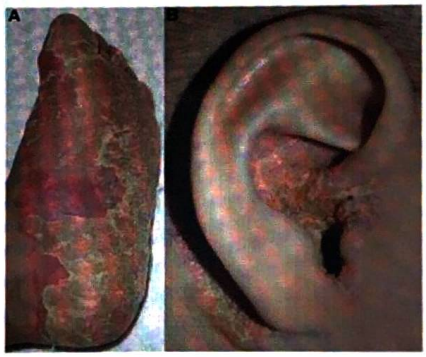


Increased levels of fumarylacetoacetate and succinylacetone leading to :

- Liver damage (chronic liver disease, cirrhosis and HCC).
  Tumor marker : Increased Alpha feto protein levels (AFP).
- Damage to the Kidney PCT : Fanconi like syndrome.

Type I tyrosinemia is AKA hepatorenal tyrosinemia.

- Recurrent acute episodes of neuropathy involving limbs: Severe pain in the limbs associated with vomiting.
- Urine odour : Boiled cabbage odour (due to accumulation of methionine metabolites).



Diagnosis :
Succinylacetone levels in the urine : Increased.

Treatment :
Reduced intake of phenylalnine and tyrosine in the diet.
Nitisinone (DOC) : Inhibits 4-HPPD (4- Hydroxyphenyl pyruvate dioxygenase)

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Tyrosinemia type 2 :
Deficiency of TAT (Tyrosine Amino Transferase).

Features :

- Corneal ulcers.
- Palmoplantar keratosis

Active space

Leave Feedba

- Keratoderma (thick, rough skin) :
- Painful, non-pruritic lesions.
- AKA oculocutaneous tyrosinemia.
- Neurological dysfunction.

Tyrosinemia type 3 :

- Least common type.
- Deficiency of 4-HPPD.
- Neurological manifestations : Developmental delay, ataxia, seizures.

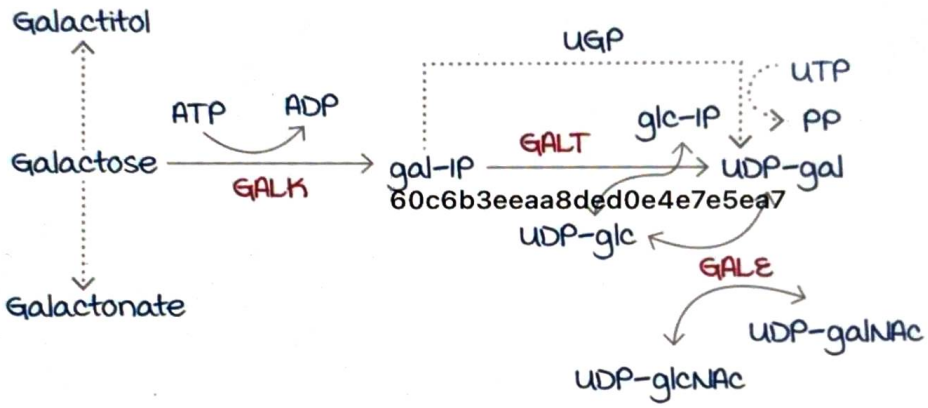
In both type 2 and 3 tyrosinemia, liver and renal functions are normal.

Galactosemia

00:56:07

Autosomal recessive.

Galactose metabolism :



GALK : Galactokinase.
GALT : Galactose-1-posphate uridylyltransferase.
UGP : UDP-glucose/galactose pyrophosphorylase.
GALE : UDP-galactose 4'-epimerase.
gal-1P : Galactose-1 phosphate.
glu-1P : Glucose-1 phosphate.

Most common enzyme deficiency : GALT.
Accumulation of galactose-1 phosphate.
Clinical features : Starts in the neonatal period after the initiation of breastfeeds (breastmilk contains lactose which can be converted to galactose).

- Galactose is not converted to glucose (hypoglycemia, seizures).
- Liver damage : Hepatomegaly, acute/chronic liver disease, cirrhosis.

Active space

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- Kidney damage : Fanconi like syndrome (PCT affected).
- Eye : Accumulation of galactitol which gets converted to dulcitol which is deposited in the lens leading to cataract (oil-drop cataract).

Cause of death : E.coli sepsis.

Late complications :
Speech defect : Delay in development of the milestone, apraxia.
Ovarian failure in female adolescents (delayed puberty).

Investigations :
Benedict's test of the urine : Positive (reducing substances).
Confirmatory test : Assay of GALT in the RBCs.

Treatment :
Eliminate galactose from the diet (breast milk is absolutely contraindicated).
No lactose formula and soy based formula is advised.
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Supplementation : Ca^{a+} and vitamin D deficiency.
Assessment of compliance is done by the levels of Galactose-1- phosphate levels in the RBCs.
- If normal : Compliant.
- If elevated : Non-compliant.

Hereditary fructose intolerance 01:05:31

Autosomal reccessively inherited.
Defect : Fructose-1 phosphate aldolase B enzyme deficiency.

Accumulation of fructose which is not converted to glucose (hypoglycemina).
Onset of symptoms : > 6 months.
Characteristic feature : Self restriction/aversion to sweets.
Liver and kidney dysfunction is seen.
No cataract formation.
Investigations : Urine analysis for fructose.

Confirmatory test : Aldolase B enzyme assay in the liver tissue.

Treatment :
- Avoid sucrose, fructose, sorbitol in the diet.
- Drugs or vitamins dispersed in sucrose base should be avoided.

Active space

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UPPER AIRWAY DISORDERS IN CHILDREN

Upper airway obstruction

00:00:20

Narrowest part of the pediatric airway in children : Sub glottic area (at the level of cricoid cartilage).

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Common feature of upper airway obstruction : Stridor (inspiratory sound).

Upper airway refers to larynx.

Conditions leading to stridor :

- Anomalies of the larynx.
- Infection in larynx.
- Foreign body in larynx.

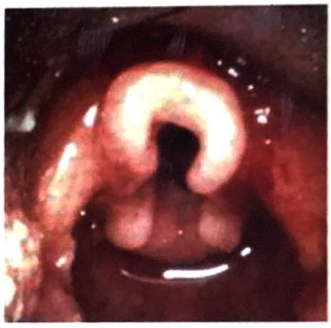
Congenital anomalies of larynx :

Laryngomalacia :

m/c anomaly of larynx.

Softness/laxity of larynx.

Structures affected : Epiglottis, arytenoids & aryepiglottic folds.



Laryngomalacia on laryngoscopy

Laryngoscopy :

Abnormality in the shape of structures in larynx → Omega (Ω) shaped epiglottis.

Increased compliance during inspiration (in supine posture) → folding (epiglottis pushed downward into airway) → obstruction of upper airway → intermittent stridor (when child is in supine position, crying/agitated , feeding).

Stridor disappears in prone posture.

Stridor : Long standing, starts soon after birth or within two weeks after birth.

Active space

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Laryngomalacia is diagnosed clinically. No investigations are needed.

Treatment :

Reassurance : Improves with age & will resolve spontaneously in 2 years.

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Severe stridor : Supraglottoplasty (rare).

Croup

00:07:16

Acute Laryngo Tracheo Bronchitis (ALTB).

Age : 6 months to 5 years (peak : 2 years).

Etiology : Para influenza virus.

Presentation : Stridor (associated with low grade fever) in an otherwise well child.

Cough : Barking/seal like/brassy cough.

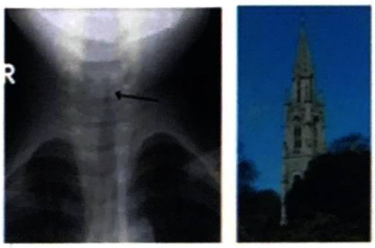
Graded based on clinical presentation :

1. Mild croup :
   - Stridor only upon coughing/exerting.
   - SpO_a : Normal.
2. Moderate :
   - Stridor at rest.
   - Respiratory distress (tachypnea/fast breathing + chest retractions).
   - SpO_a : Normal.
3. Severe :
   - Stridor at rest.
   - Respiratory distress (tachypnea/fast breathing + chest retractions).
   - Hypoxia (SpO_a < 92 % in room air).

X ray :

Narrowing of airway (maximum narrowing in sub glottic area) :

Steeple sign.



Steeple sign

Active space

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Management :

Mainly supportive treatment.

1. Mild croup : Oral Dexamethasone (0.15 – 0.6 mg/kg) as single dose.
2. Moderate or severe croup :

   If hypoxia is present : O_2 inhalation & nebulization with racemic epinephrine (racemic : D & L isomers of epinephrine) or L epinephrine alone (whichever is available).

Acute epiglottitis

00:14:11

It is a medical emergency.

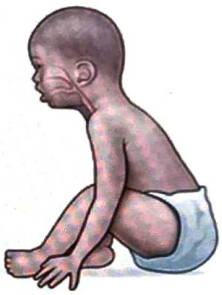
Age group : 1 to 3 years (toddler age group).

Etiology : H. influenza type B (most common).

Presentation :

Sudden onset high grade fever.
Sick (toxic) looking.
Stridor.
Drooling of saliva.
Respiratory distress.

Characteristic posture : Tripod posture/sniffing dog posture.



Tripod position : Epiglottitis

Investigations :

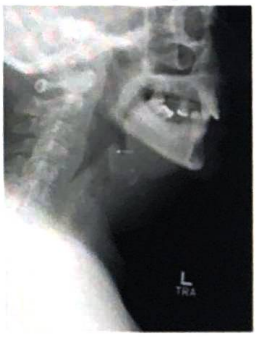
1. Lateral X ray of neck :

   Thumb sign (swollen epiglottis causes narrowing of airway → gives impression of thumb).

2. Laryngoscopy : Cherry red epiglottis (severely inflamed epiglottis).

   Laryngoscopy is not done in acute epiglottitis because of risk of acute laryngeal spasm while maneuvering.

Thumb sign



Management :

1. Priority : Secure airway (endotracheal intubation or tracheotomy).

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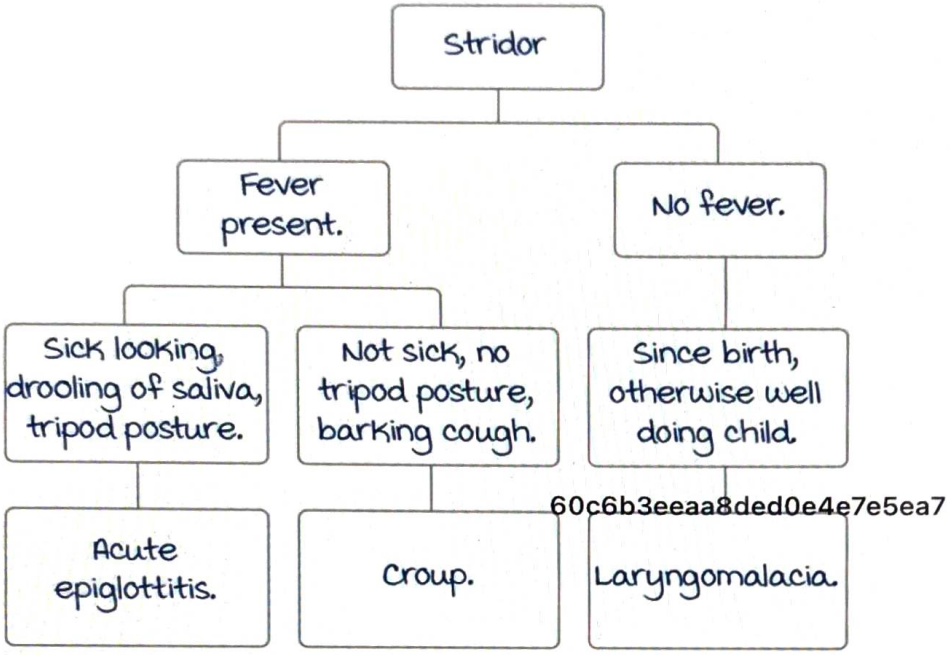
2. Antibiotics : 3rd generation cephalosporin (Ceftriaxone or Cefotaxime).
3. No role for sedatives.

Differentials for stridor

00:20:50



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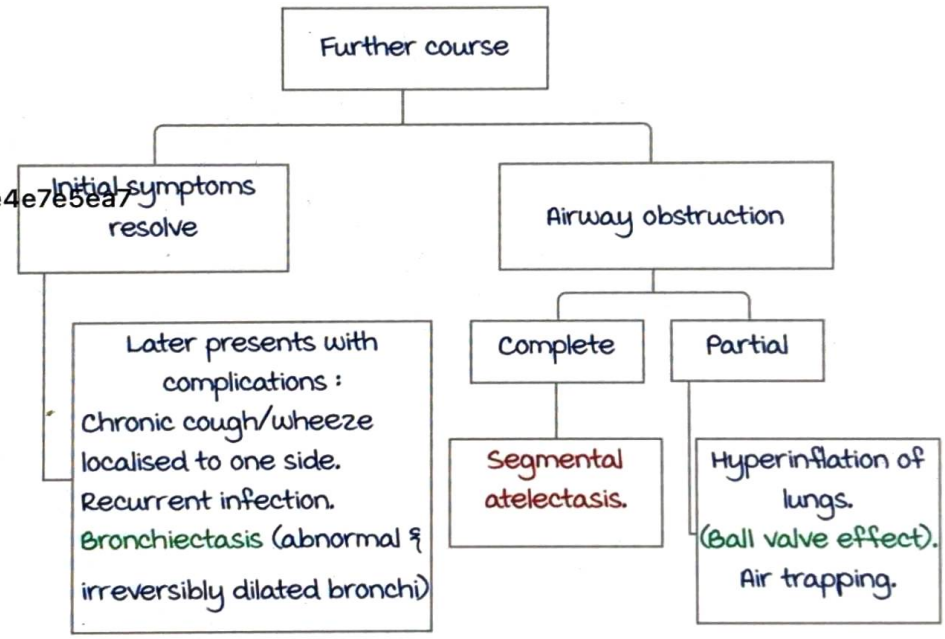
FOREIGN BODY ASPIRATION, CONGENITAL LUNG ANOMALIES & ASTHMA

Foreign body aspiration

00:00:13

Commonly seen in age < 2 years.
Most common foreign bodies : Peanuts, coins.
Lower airway foreign body aspiration :

- Location : Bronchus (right and left bronchus equally affected).
- Presentation :
  Sudden onset cough/choke/gag/breathing difficulty.
  Presents without fever.
- Most common presentation : Sudden onset of Cough.
- Upon auscultation, unilateral wheeze is heard.



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Air cannot pass through a completely blocked bronchus → No inflation of the respective lung segment → Segmental atelectasis.

Ball valve effect : During inspiration, air goes in.
During expirartion, no air goes out.

Active space

Chronic presence of foreign body is suspected when a child has recurrent pneumonia involving the same side of lungs & also the same segment.

Only 10 – 20% of foreign bodies are radio opaque (all foreign bodies may not be seen on a X ray).

Suspicion of foreign body aspiration 00:07:26

1. History suggestive of sudden onset symptoms.
2. Focal abnormal lung findings.
3. Abnormal X ray.

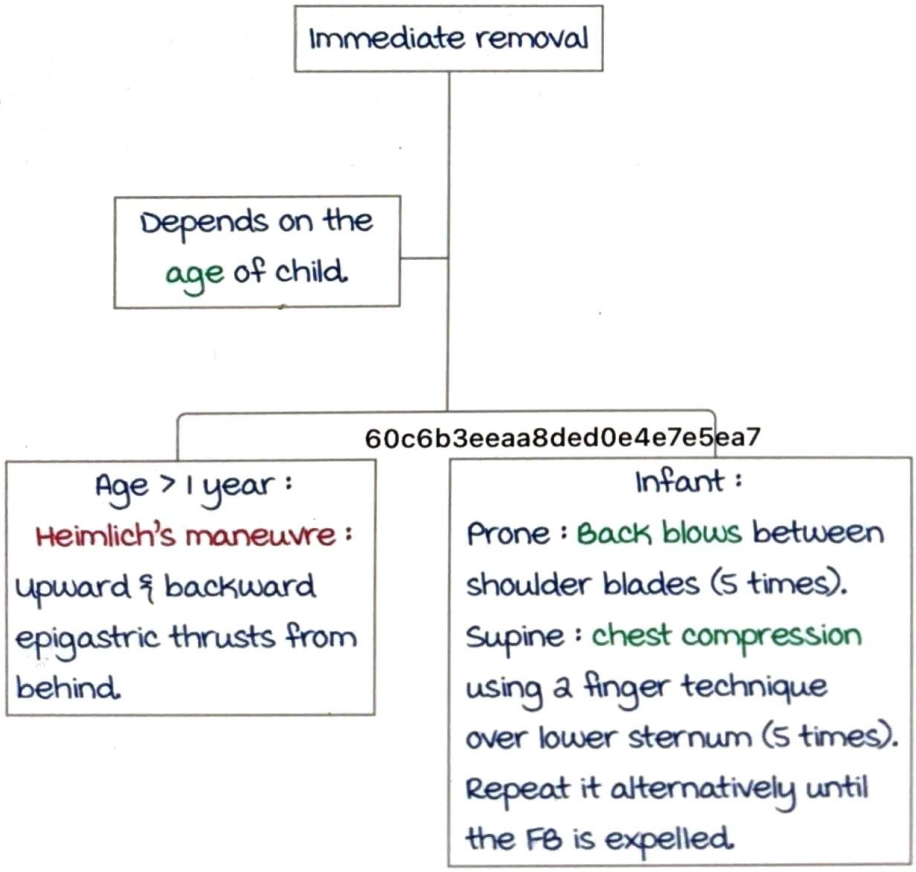
If any 2 out of 3 present, the suspicion is strong and removal of the foreign body is initiated.

Treatment : Rigid bronchoscopy guided removal under GA.

Upper airway foreign body(FB) aspiration :

Presentation : Stridor.

Treatment :



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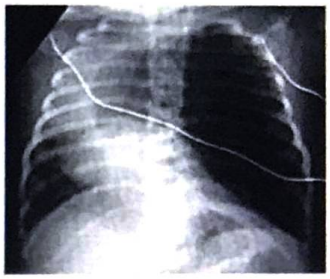
Congenital Lung anomalies :

Congenital lobar emphysema :

- Also called as congenital lobar hyperinflation.
- Cause : Cartilaginous bronchial dysplasia (developmental defect) leading to partial ball valve effect and result in air trapping and hyperinflation.
- Site : Left upper lobe.
  Right middle lobe.
- Characteristic features :
  Mediastinal shift.
  Respiratory distress in newborn.
- IOC : CT chest.
- Treatment : Lobectomy.



Pulmonary sequestration

00:14:51

Non functional pulmonary tissue that does not communicate with tracheobronchial tree.
Blood supply from systemic arteries.
Classified as intralobar or extralobar.
Extra lobar has separate pleural covering. Intra lobar is covered by common pleura.
Site : Lower lobes of Left lung
Complication : Recurrent respiratory infection.
IOC : CT Chest.
Treatment : Surgical resection.

Congenital Pulmonary Adenomatoid Malformation (CPAM) :
Previously called as CCAM (Congenital Cystic Adenomatoid Malformation).

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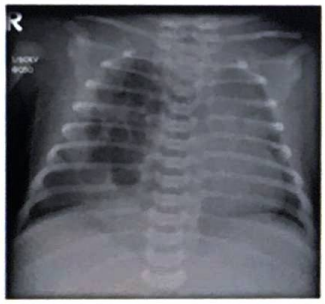
MC cause of cystic lung disease in newborn.
Unilateral hamartomatous lesions.
Site : Left lung.
Presents either in newborn or in older children as recurrent infection.
Sometimes, it is asymptomatic.
Risk of malignancy (either sarcoma/carcinoma) of lung.
Treatment : Surgical resection within one year of age.



Active space

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Asthma

00:20:02

Characterized by bronchoconstriction.

Present with wheeze (airway narrowing).

Pre requisites to diagnose asthma :

- Triggered by allergens or infections.
- Reversible (either spontaneously or using short acting beta agonists).
- Recurrent episodes (at least 3 or more)
- Presence of risk factors like
    1. History of asthma/other allergies in parents.
    2. Other allergic disorders like atopic dermatitis, sinusitis, allergic rhinitis.
    3. Male gender.
    4. Low socio economic status.
    5. Repeated exposure to tobacco smoke.

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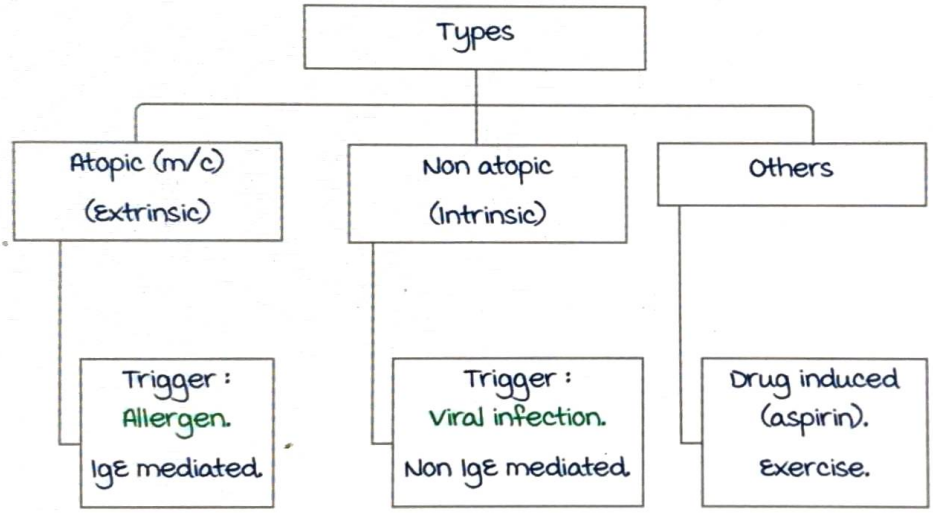
Triggers of asthma include : Dust, pollen, exercise, stress & respiratory infections.



Cough variant asthma :

- Presents with recurrent cough.
- Seen in young children.

Investigations to diagnose asthma :

Spirometry :

1. Evidence of airflow limitation : Low FEV1, FEV1/FVC < 0.8
2. Bronchodilator response : Increased FEV1 ≥ 12% after inhalation of bronchodilator like salbutamol.

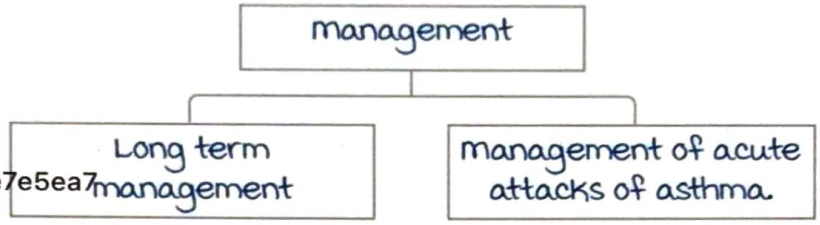
Active space

Leave Feedback

3. Exercise challenge : Worsening FEV1 ≥ 15% after exercise.
4. Daily PEF or FEV1 monitoring : AM-PM variation and/or day to day variation (PEF : Peak Expiratory Flow). Variation ≥ 20% is suggestive of asthma.
5. Exhaled nitric oxide : >20 ppb (parts per billion).

Management of asthma

00:28:14



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Long term management : Step wise management :

1. Intermittent asthma is characterized by :
   - Infrequent symptoms.
   - Child is normal in between attacks.

   Treatment : Short acting beta agonists (as and when needed).

2. Persistent asthma :
   - Mild persistent : Day episode ≥ 2/month.
     Night time awakening ≥ 1/month.

     Treat daily with low dose inhaled corticosteroids (ICS) : Budesonide, Fluticasone, Beclomethasone.

   - Moderate persistent :

     Daily attacks or need to use of SABA daily.

     Treat with medium dose ICS or low dose ICS daily + LABA (Long Acting Beta Agonist) like Salmeterol, formoterol.

   - Severe persistent :

     Daily attacks but continuous throughout day & night.

     Significant limitation of physical activities.

     Treatment : High dose ICS daily + LABA.

Alternate choices (as add on drugs for better control) :

- Montelukast.

Active space

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- Sustained release theophylline.
- Oral short course steroids or alternate day steroids.

Oral short course streoids are used only when asthma fails to respond to every other drug (to avoid systemic complications).

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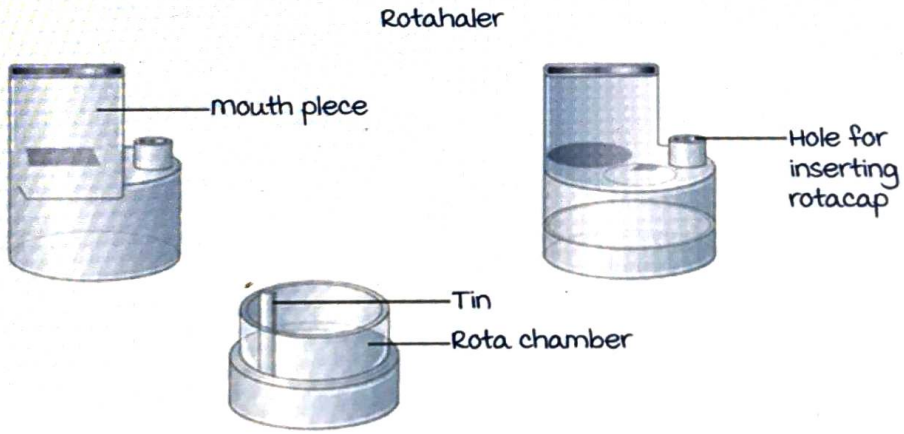
Asthma medication devices 00:36:18

- metered dose inhaler (MDI) :
  Given along with a spacer (small amount of drug goes in with every puff).
  < 4 years : Face mask + Spacer+ MDI.
  4–12 years : Spacer + MDI.
  > 12 years : Only MDI.

- Rotahaler :
  Also called dry powder inhaler (DPI) : > 5 years.



Rotacap is a capsule containing the drug.

Nebulizer can be used to administer the medicines in a hospital setup.

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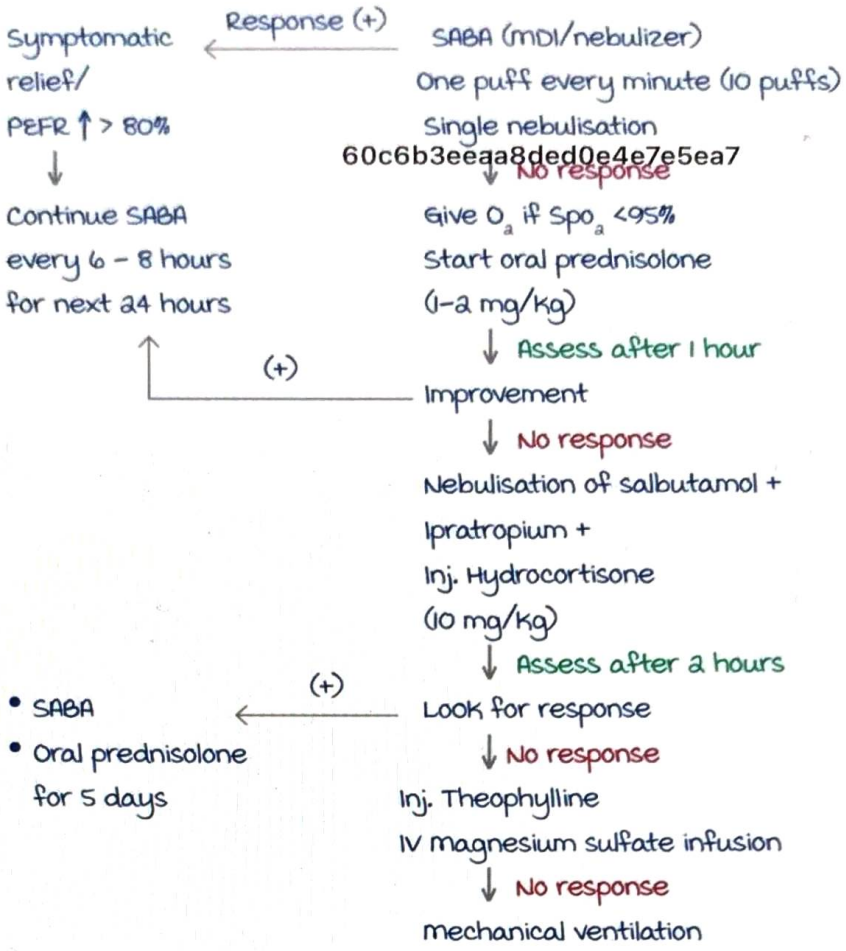
Management of acute attacks of asthma :



Tapering of oral prednisolone is not required.
Oral steroids followed by IV steroids is the norm.

Life threatening asthma

00:46:49

Any child presenting with :

- Silent chest.
- Cyanosis.
- Altered sensorium.
- Poor respiratory efforts/laboured breathing.
- SpO2 < 90%.
- PEFR < 30% of expected.

Treatment :

- Injection Adrenaline/Terbutaline subcutaneously.
- Injection Hydrocortisone.
- Neb. Salbutamol + Ipratropium.

Active space

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LOWER RESPIRATORY TRACT INFECTIONS

Acute bronchiolitis

00:00:21

It is a common seasonal infection which usually occurs in the winter and spring seasons.

most common cause : RSV (Respiratory Syncytial Virus).

Age group : < 2 years (mostly in 1 to 6 months of age).

Clinical presentation :

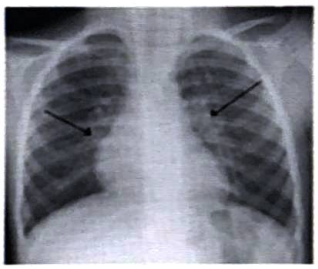
Initially starts as an upper respiratory tract infection.

After 1 week, it progresses on to the lower respiratory tract (bronchioles) leading to :

- Fever
- Breathing difficulty
- B/L wheeze (due to passage of air through narrowed and inflamed bronchioles)

X ray findings :

- B/L hyperinflation of the lung fields. The bronchioles are inflamed and are hence narrowed.

  During inspiration, some amount of air will be able to pass through bronchioles as they are not fully obstructed.

  But during expiration, air columns tend to become smaller and there is almost complete obstruction. This results in trapping of air inside the alveoli of the affected bronchioles.
- Perihilar fullness/perihilar cuffing (due to inflammation of bronchioles in the hilar regions : White markings).

Clinical findings due to hyperinflation :

- Increased AP (antero-posterior) diameter of chest.
- Hyper resonance on percussion.

Clinical findings + X-ray is enough to make diagnosis.

Other investigation : PCR testing of the nasopharyngeal swab /aspirate (not done clinically).

Active space

Leave Feedba

Treatment :

It is usually a self-limiting condition.

Supportive management :

- moist O_a inhalation to maintain SpO2 > 92% in room air.
- Fluids (oral or iv fluids).

Antivirals are prescribed for at risk children.

- Chronic lung diseases.
- Congenital heart defects.
- Immunodeficient children.

Antivirals prescribed are :

- DOC : Ribavirin (inhalational form) - decreases the duration and severity of the illness.
- Before the start of the season, prophylactic Palivizumab is given (monoclonal antibody against F protein of RSV).

Bronchiolitis usually subsides within 3-7 days.

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25% of the patients with bronchiolitis develop asthma in future.

Pneumonia

00:10:36

It is an important cause of under 5 mortality.

Definition : Pathological consolidation of alveoli or inflammation of the interstitial tissue around alveoli or both.

Aetiology : Bacterial > Viral.

- Overall most common bacteria : Streptococcus pneumoniae.
- Most common cause in neonates : Gram negative organisms (Klebsiella, E.coli), Staphylococcus aureus.
- Most common virus : RSV (Respiratory Syncytial Virus).
- In children with HIV : Pneumocystis jiroveci (carinii).
- In adolescents/older children (> 5 years) atypical age group : mycoplasma pneumoniae (atypical virus).

Pneumonia is an important public health problem because of under 5 mortality.

Active space

Leave Feedbac

Community management guidelines 00:13:44

Revised WHO – IMNCI guidelines :

Category	Features	Management
No pneumonia. Color code : Green.	Fever, cold, cough.	Home management : Oral paracetamol. If wheeze present : Salbutamol for 5 days.
Pneumonia. Color code : Yellow.	Fever, cough, cold + fast breathing and/or chest indrawing. SpO_a normal (no hypoxia)	Home management + oral antibiotic (Amoxicillin) x 5 days. Follow up after 5 days. Look for danger signs.
Severe pneumonia/Very severe disease. Color code : Pink.	Any one of the following should be present : • Hypoxia : SpO2 < 90% in room air. • Stridor in a calm child. • If any danger signs are present : 1. Not feeding/drinking 2. Lethargic. 3. Cyanosis all over body. 4. Convulsions.	Give the first dose of antibiotic (Ampicillin + Gentamicin). Immediately refer the child to the hospital.

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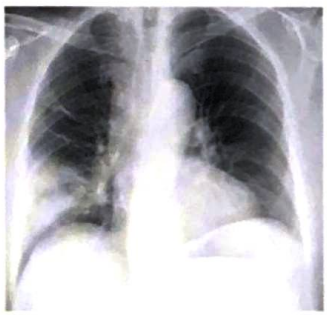
Criteria for fast breathing :

Age	Respiratory Rate
0 – 2 months	> 60/minute
2 – 12 months	> 50/minute
> 12 months	> 40/minute

X-ray patterns of pneumonia 00:20:54

Streptococcus pneumoniae :
X – ray finding : Lobar consolidation.
It is associated with typical rusty sputum (not seen as children swallows sputum)



Lobar pneumonia

Active space

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Treatment :

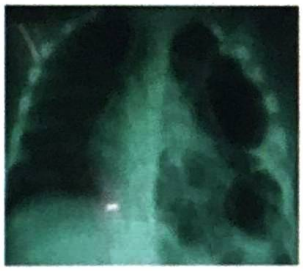
- Penicillin G 50,000 IU/kg/day IV.
- Co-amoxiclav.
- 3rd generation cephalosporins (Ceftriaxone, Cefotaxime). Used nowadays because of resistance to amoxicillin.

Staphylococcus aureus :

X - ray pattern :

- B/L patchy consolidation.
- Pneumatoceles (air filled spaces). Can rupture & lead to pneumothorax.

Staphylococcal pneumonia



Risk factors :

- Severe malnutrition
- Diabetes.
- As a complication of influenza, measles infection.

Complications :

- Pneumothorax.
- Empyema (pus in pleural cavity).

It is a severe pneumonia in children.

Treatment :

Penicillin-G/Co-amoxiclav.

If no response, add 3rd generation cephalosporins.

If MRSA is suspected : Add Vancomycin/Linezolid.

Conditions in which pneumatoceles are seen :

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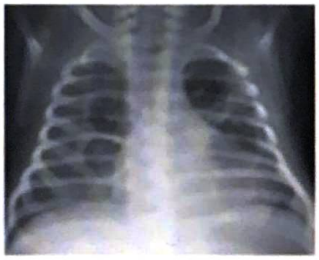
- Staphylococcus aureus associated pneumonia.
- Klebsiella/E. coli associated pneumonia.
- Hydrocarbon poisoning.



Pneumatocele

Active space

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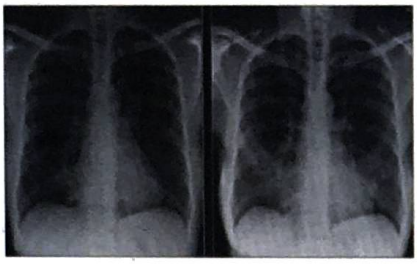
Mycoplasma pneumonia or RSV (Atypical pneumonia) :
X ray pattern : B/L interstitial pneumonia
(bronchopneumonia).

M. pneumoniae features :
Usually affects older children (> 5 years).
The child appears clinically well (walking pneumonia)
(Lot of interstitial pattern but a well child)
Treatment : Macrolides (Azithromycin).



Interstitial pneumonia

Recurrent pneumonia and persistent pneumonia

00:31:12

Recurrent pneumonia : ≥ 2 episodes in 1 year or ≥ 3 episodes in a lifetime.
Persistent Pneumonia : Non-resolving pneumonia, characterised by persistent radiological findings for > 1 month.

Risk factors for recurrent/persistent pneumonia :
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- Severe malnutrition.
- Immunodeficiency.
- Recurrent aspiration :
    1. GERD.
    2. Upper GI anomalies like esophageal atresia (aspiration of vomitus).
- Congenital heart defects (due to pulmonary congestion).
- Genetic disorders :
    1. Cystic fibrosis.
    2. Kartagener's syndrome.

If risk factors are treated, recurrence could be avoided.

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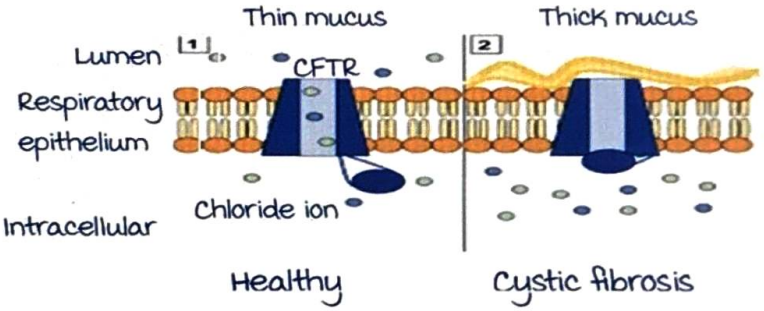
CYSTIC FIBROSIS

Introduction to cystic fibrosis (CF) 00:00:21

- Cystic fibrosis is a multi-sysem AR disorder (GIT, RS, Genito-urinary, sweat glands are affected).
- One of the most common life limiting genetic disorder.
- Seen mostly in western population. Cases continue to increase in India as well.

CFTR gene (CF transmembrane regulator) → located on chromosome 7q → codes for CFTR protein (Cl^- channel).
Mutation of CFTR gene causes cystic fibrosis.
m/c mutation : ΔF 508 → deletion of phenylalanine at 508th position. > 1500 mutations have been described in the gene.

Mucoviscidosis :
Thick mucus secretions in body.



Respiratory epithelium :
In normal people :
CFTR channel → Cl^- ions transported from intracellular portion into lumen → Cl^- makes the mucus thin → thin mucus easily expelled out (mucociliary clearance).

Cystic fibrosis :
Inactive CFTR channel → Cl^- ions cannot be transported into lumen → thick mucus → mucus cannot be expelled out → mucoviscidosis → recurrent infections of respiratory tract (recurrent pneumonia).

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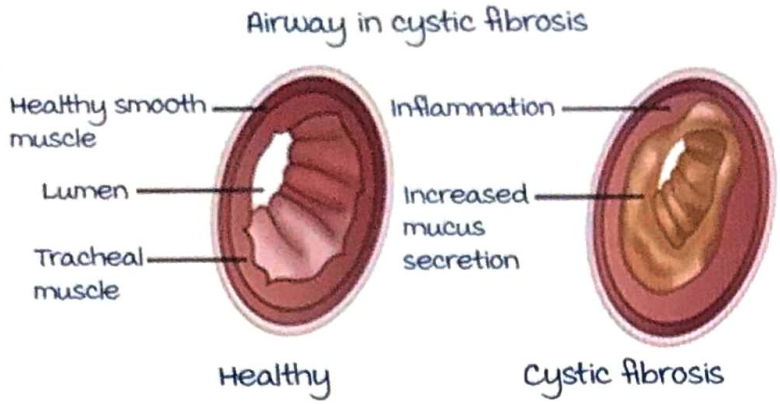


Clinical features

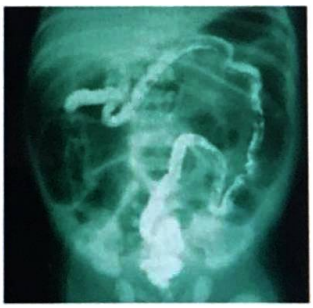
00:05:54

Gastrointestinal tract : Earliest features.

1. Meconium ileus : Normally meconium is the 1st stool passed by a newborn baby from small intestinal secretions.
   - Early manifestation.
   - In cystic fibrosis → meconium becomes thick → not easily expelled out → stuck in intestinal lumen → inspissation of thick meconium in ileum (intestinal obstruction at ileum) → patient presents with delayed passage of meconium & abdominal distension.
   - Virtually diagnostic of CF but present in only 10 – 15% patients.

On contrast enema :

- Microcolon (large intestine appears small) + filling defects in colon.
- Obstruction at terminal ileum : Distended small intestine, proximal to terminal ileum + air fluid levels.
- In lower part of abdomen, ground glass appearance (due to meconium + air causing opacification) on plain X-ray is seen.



2. Constipation (occurs in older children) :
   Impaction of fecal matter in distal part of ileum : DIOS (Distal Intestinal Obstruction Syndrome).

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3. Exocrine pancreatic insufficiency :

Pancreatic secretions become thick → not expelled into duodenum → fat and fat soluble vitamins (Vit A, D, E, K) are not absorbed → steatorrhea + deficiency of vit A, D, E, K.

85 % of the children have this condition.

one of the m/c manifestation of cystic fibrosis.

4. Endocrine pancreatic insufficiency :

In 2nd decade → Endocrine pancreatic insufficiency → decreased insulin secretion → may cause Diabetes mellitus in future.

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5. Rectal prolapse :

Uncommon due to early supplementation pancreatic enzyme. Presents, if at all, in older children.

Respiratory tract manifestations :

1. Recurrent infections :

Different organisms causing infection or pneumonia in a child with cystic fibrosis :

- In early childhood : Staphylococcus aureus (overall m/c), H. influenza type B.
- In late childhood/adults : Pseudomonas.

Pseudomonas colonizes the respiratory tract & produces mucoid secretions.

Forms biofilm around the organism → provides antibiotic resistance to Pseudomonas.

- Burkholderia cepacia : Increased risk of death in cystic fibrosis.

2. Nasal polyps :

In a child with bilateral nasal polyps → cystic fibrosis should be evaluated.

Other features of cystic fibrosis 00:16:14

Biliary tract :

Biliary cirrhosis/neonatal cholestasis (thickening of biliary secretions → causing bile outflow obstruction).

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Pancreatic insufficiency (initially exocrine → endocrine).

Genitourinary tract manifestations :
All affected males → infertile → associated with azoospermia (failure of development of wolffian duct).
Females → decreased fertility rate.

Sweat glands :
Normally, CFTR protein helps in the reabsorption of Na^+/Cl^- from sweat glands.
In cystic fibrosis : CFTR protein non-functional.

↓

Excessive loss of Na^+/Cl^- in sweat.

↓

Salty skin (may feel salty while kissing).
Frosting of skin.

Predisposed to hyponatremic hypochloremic metabolic alkalosis.
Increased sweat Cl^- helps in diagnosis.

Diagnostic criteria for cystic fibrosis 00:19:49

When to suspect?

- Presence of typical clinical features : Respiratory, gastrointestinal/genitourinary.
- History of CF in a sibling as its Autosomal recessive.
- Positive newborn screening test : Immunoreactive trypsin test (IRT). Commonly done in western countries.

Presence of any 1, CF should be confirmed with lab tests.

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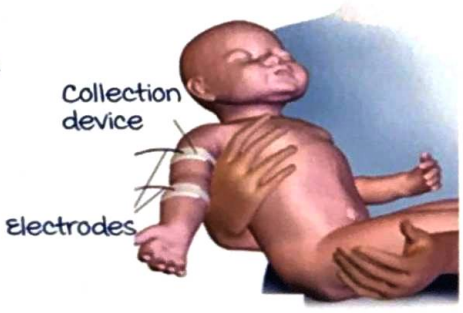
Confirmation (lab evidence) :

- 2 elevated sweat chloride concentrations (>60 mEq/L) obtained on separate days.



Sweat test

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Pilocarpine iontophoresis :
Pilocarpine is administered into skin of baby with help of electrodes.
↓
Baby's sweat will be collected in collection device.
↓
The collection device will automatically process the levels of Cl^-.
(or)

- Identification of two CF mutations (or)
- An abnormal trans epithelial nasal potential difference measurement : Single abnormal value is sufficient.

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If any 1 lab test is positive, diagnosis of CF is made.

Management of cystic fibrosis 00:23:30

Respiratory care :

- Airway clearance treatment :
  Physiotherapy (to remove thick mucus).
  Mucolytics : Inhaled hypertonic saline (3%).
  Human recombinant DNAse (human Dornase alfa).
- Antibiotics :
  1. Co-amoxiclav (Amoxicillin and Clavulanic acid) : Preferred as first antibiotic.
  2. Fluoroquinolone.
  3. Azithromycin.
- Prophylaxis for Pseudomonas infection :
  Inhaled Tobramycin or inhaled Aztreonam (or) oral Azithromycin 3 times a week (decreases colonization).

Dietary measures :

- Diet : High fat, protein, calories.
- Salt supplementation (more during dehydration).
- Pancreatic enzyme supplements (enterically coated microspheres). It is the cornerstone of gastrointestinal management in cystic fibrosis.

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- Fat soluble vitamin supplementation (A, D, E & K):
  Vit A & D are the most important supplements.

CFTR modulators:

Increases the activity of CFTR channel by modulating it.

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1. Ivacaftor: > 2 years of age.
2. Ivacaftor + Lumacaftor: > 6 years of age.
   useful in ΔF 508 mutation cases.
3. Ivacaftor + Tezacaftor: > 12 years of age.
4. Triple combination therapy:
   Ivacaftor + Tezacaftor + Elexacaftor.
   Covers most of the mutations (most promising evolving treatment).

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FETAL CIRCULATION & INTRO TO CONGENITAL HEART DISEASES

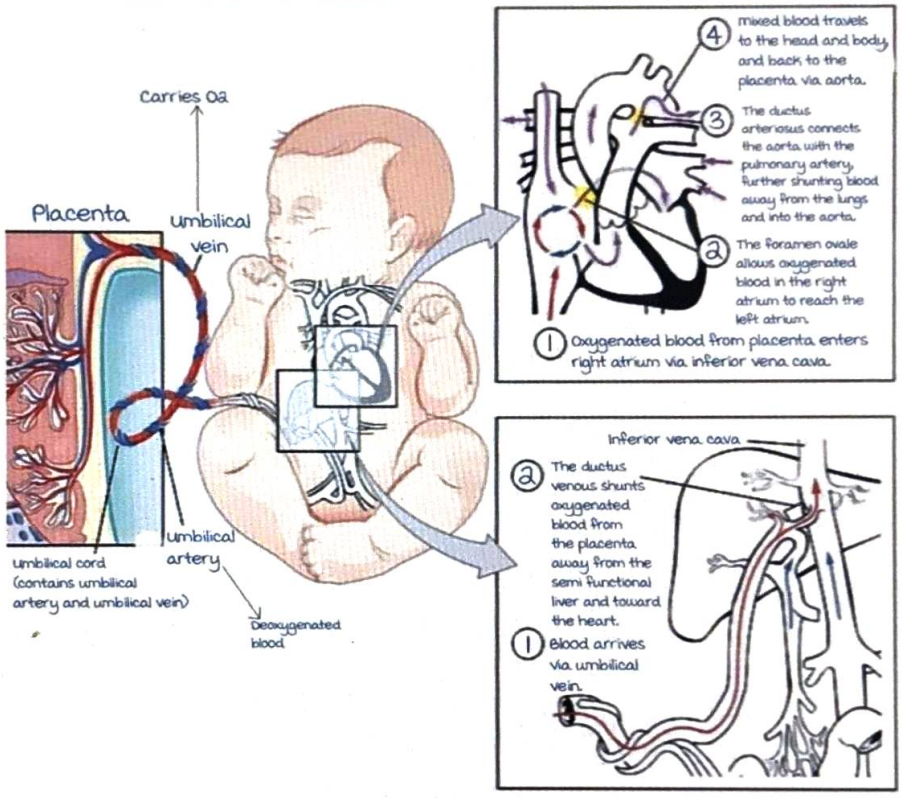
Fetal circulation

00:00:26

- Placenta provides oxygen (by umbilical vein) and nutrients to the baby as the lungs aren't yet functional.
- High pulmonary vascular resistance.

Special structures :

- Ductus venosus.
- Foramen ovale.
- Ductus arteriosus.



The umbilical artery carries deoxygenated blood.
The umbilical vein carries oxygenated blood. The ductus venosus connects the umbilical vein to IVC and bypasses the liver.

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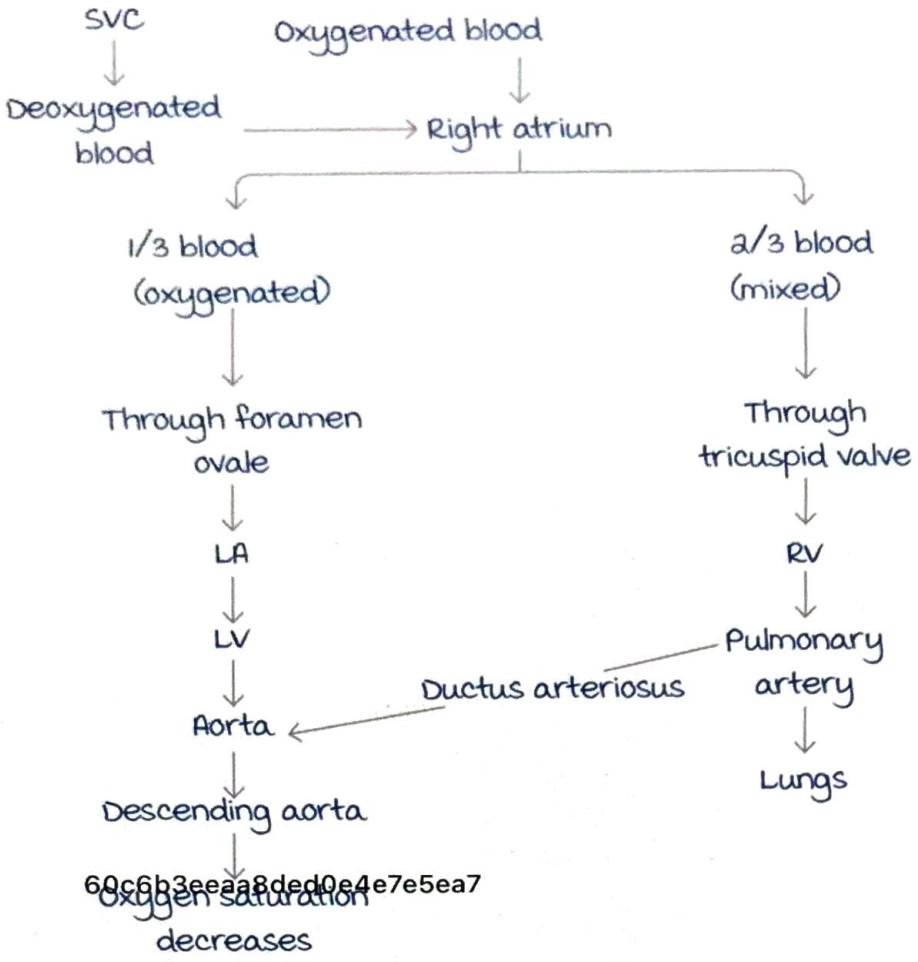

	Oxygen saturation
Umbilical vein	80 %
Ductus venosus	75 %
IVC	70 %
LV	70 %
RV	55 %
Descending aorta	55 % (50 – 60 %)
Umbilical artery	55 % (50 – 60 %)

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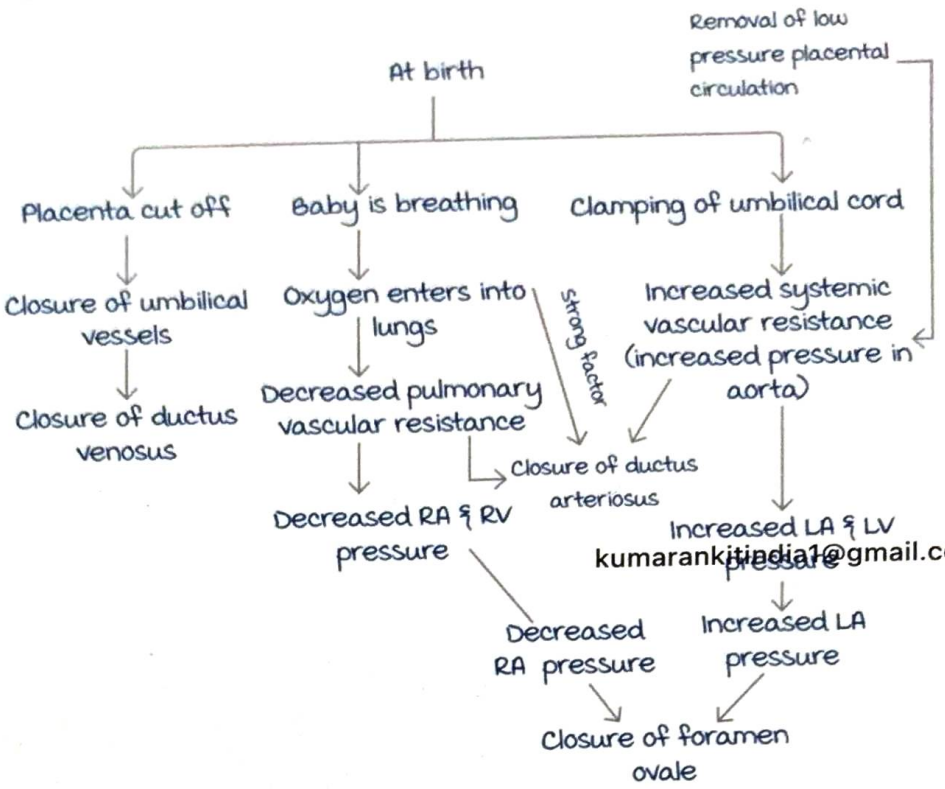
Circulatory changes at birth

00:09:56



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Structure	Functional Closure	Structural closure after birth (anatomical)	Remnant
Umbilical vessels	At/soon after birth	5 – 10 days	Umbilical Vein : Ligamentum Teres. Umbilical Artery : medial umbilical ligament.
Ductus venosus	At/soon after birth	3 – 7 days	Ligamentum Venosum.
Foramen Ovale	At/soon after birth	3 months up to few years (structurally patent foramen ovale).	Fossa ovalis.
Ductus arteriorus	10 to 14 hours after birth	10 –21 days	Ligamentum arteriosum.

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Timeline questions can be answered on the basis of functional closure.

Last structure to functionally close : Ductus arteriosus.
Last structure to close anatomically : Foramen ovale.

Congenital heart diseases/CHD 00:20:32

Prevalence : 6 to 8/1000 live births.
Polygenic/multifactorial inheritance.
MC CHD : Ventricular Septal Defect (VSD).
MC cyanotic congenital heart disease : Tetralogy Of Fallot (TOF).

CHD associations :
Genetic syndromes :

- Down syndrome (trisomy 21) : Endocardial cushion defect.
  1. Atrio-ventricular septal defect.
  2. AV valve defects (most commonly regurgitation).
- Turner syndrome :
  1. Bicuspid aortic valve.
  2. Coarctation of aorta.
- Noonan syndrome :
  1. Pulmonary stenosis.
  2. ASD.
  3. HOCM (Hypertrophic Obstructive Cardiomyopathy).
- Edwards syndrome (trisomy 18) & Patau syndrome (trisomy 13) : VSD.
- Digeorge syndrome : Conotruncal defects.
- Williams syndrome : Supravalvular aortic stenosis.
- Holt Oram syndrome (AD) : Skeletal anomalies (Absent radius m/c) + ASD (strongest familial association).
- Alagille syndrome : Pulmonary stenosis.

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Prenatal (maternal) exposure :

- Diabetes (overt) : VSD.
- Congenital Rubella syndrome :
  1. Patent ductus arteriosus.

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2. Pulmonary stenosis.
3. VSD.

- Lupus : Anti-Ro (SS-A)/anti La (SS-B) → Antibodies cross placenta Congenital heart block in fetus.

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- Teratogens :
  1. Alcohol & Phenytoin : VSD.
  2. Lithium : Ebstein's anomaly.

NADA's criteria for CHD

00:30:24

CHD is possible if one major or 2 minor criteria are present.

Major criteria :

- Systolic murmur : Grade 3 or more.
- Diastolic murmur : Any grade.
- Cyanosis.
- CCF.

Minor criteria :

- Systolic murmur < grade 3.
- Abnormal S2
- Abnormal chest X ray.
- Abnormal BP.
- Abnormal ECG.

Diagnostic considerations :

1. Systolic murmurs :

Pansystolic murmur :

- Heard throughout systole.
- They are high grade murmurs (grade 3 or more).
- Always pathological.
- Seen in : VSD, mitral regurgitation & tricuspid regurgitation.

Ejection systolic murmur :

- Pathological : Grade 3 or more, heard when blood gets ejected out.
  Seen in aortic stenosis, pulmonary stenosis.
- Normal : < grade 3, also called short systolic murmur.
  Seen in high fever & anemia.

Active space

a. Cyanosis : Results from severe hypoxia (SpO_2 < 85%).
(Hypoxia < 94%)

	Central Cyanosis	Peripheral Cyanosis
Reason	Associated with CHD. Cyanosis due to : 1. Mixing (right → left). 2. Obstruction to pulmonary blood flow.	Due to conditions leading to vasoconstriction of limbs → Decreased oxygen delivery to tissues.
Site	Mucus membranes. Extremities.	Extremities (hands/feet).

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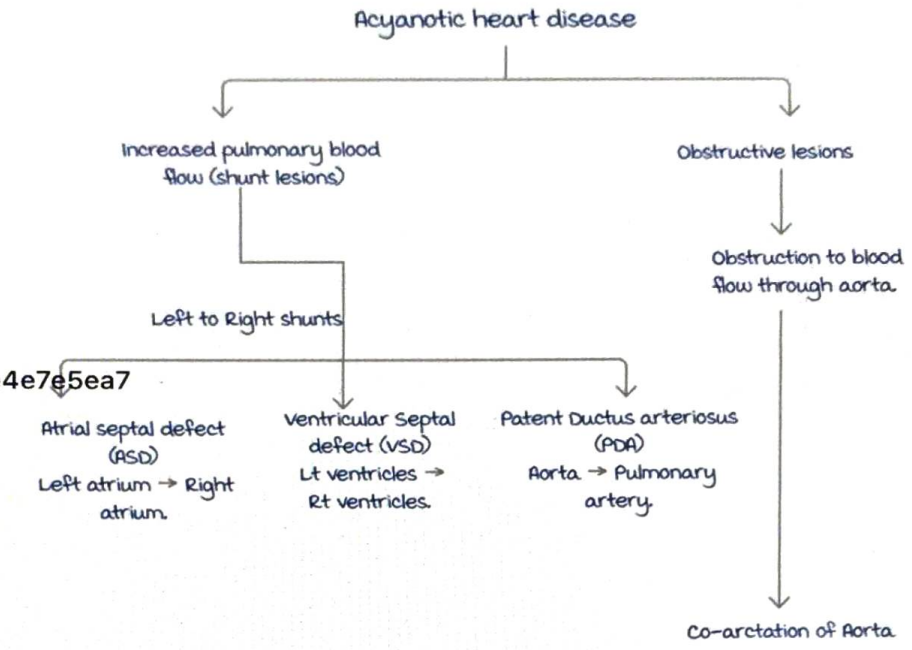
ACYANOTIC CONGENITAL HEART DEFECTS

Acyanotic CHD >> cyanotic CHD.



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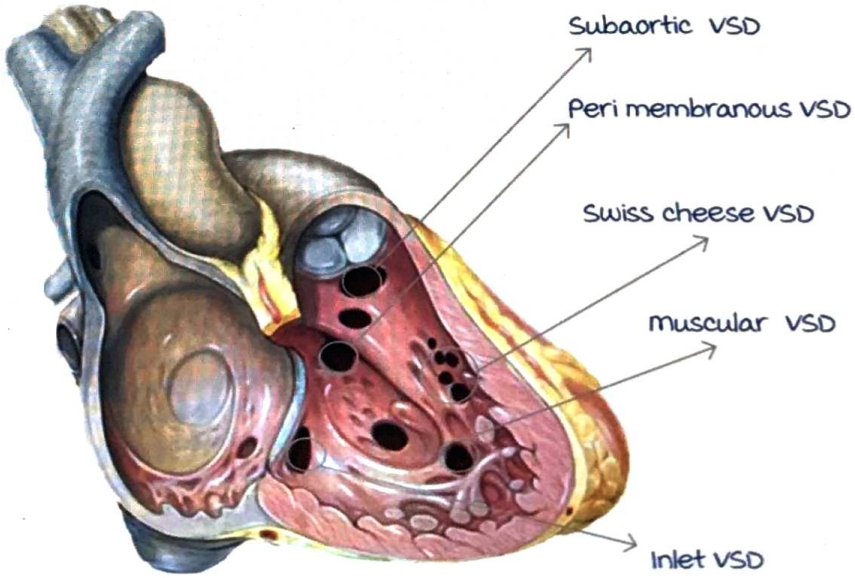
Ventricular septal defect / VSD

00:03:50



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MC overall congenital cardiac defects (25%).

Types :

- Peri membranous VSD : Overall MC type of septal defect (90%).
- Muscular VSD : High chance of spontaneous closure.
  Swiss cheese VSD (special type) : Low chance of spontaneous closure.
- Sub aortic/ outlet/ supra cristal VSD (never close spontaneously) : Associated with increased risk of aortic regurgitation.
- Inlet VSD : Associated with increased risk of mitral regurgitation.

Pathophysiology :

Whenever blood flows from left ventricle (high pressure) to right ventricle (low pressure), turbulence is caused → Pansystolic murmur heard over the left 4th intercostal space/ parasternal area (classical murmur).

Also called as Shunt murmur or Roger's murmur.

Flow murmurs :

- Ejection systolic murmur in the pulmonary region : Increased volume of blood passes from right ventricle to pulmonary artery through pulmonary valve.
- Delayed diastolic murmur in the mitral region.

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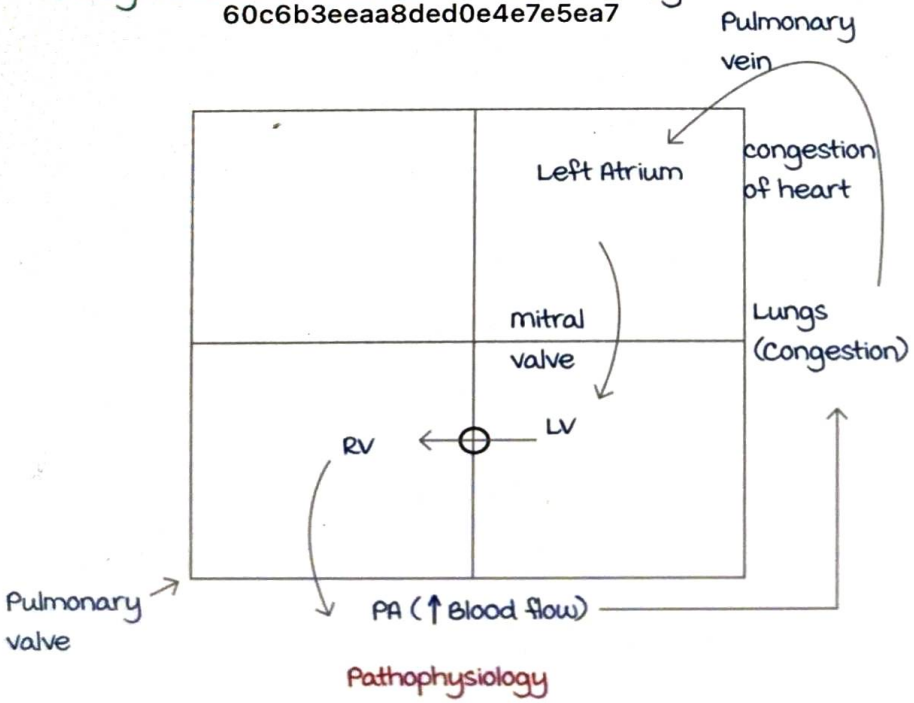

Pathophysiology

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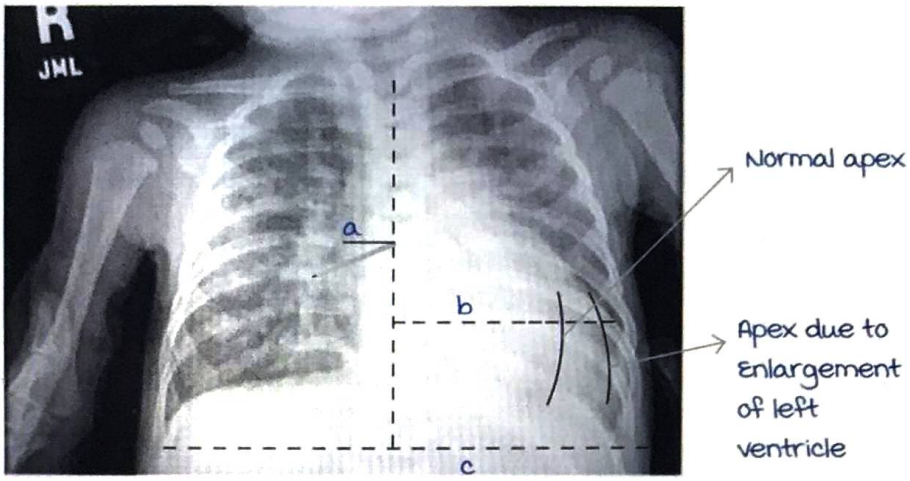
Due to overloading in VSD there is an enlargement of left ventricle → Downward and outward pushing of the apex. (characteristic feature).

Congestion of heart is seen as cardiomegaly.

Cardiothoracic ratio = (a+ b)/c.



a + b → Longest cardiac diameter.

c → Longest thoracic diameter.

cardiothoracic ratio > 0.55 → In older children, 0.6 in infants.

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Complications :

- Lung congestion : Recurrent pneumonia.
- Congestive cardiac failure / CCF : Occurs around 6 -10 weeks after birth which coincides with pulmonary vascular resistance (PVR) being very low .
- Infective endocarditis → MC associated complication of VSD.
  MC cause of infective endocarditis in children : Streptococcus viridans.
- Late complication : Eisenmenger's syndrome :
  Constant increased blood flow in pulmonary artery (for few years) → Causes irreversible changes (due to endothelial dysfunction + vascular remodelling) → Leading to increased pressure /narrowing of the pulmonary artery → Right ventricular hypertrophy (RVH).
  As the pressure increases in the right ventricle (deoxygenated) the blood starts flowing into the left

Active space

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ventricle (oxygenated) through VSD → Reversal of shunt known as Eisenmenger's syndrome.
Aorta gets mixed blood from the LV → Cyanosis.
Features → Cyanosis in a previously acyanotic patient, clubbing etc.

Treatment :
Medical : To Control CCF : Digoxin, Furosemide.
To treat infections : Broad spectrum antibiotics.
Surgical closure indications :

- Features of CCF.
- Q_p (pulmonary blood flow) : Q_s (systemic blood flow) ratio >2 : 1.
- Supracristal VSD.

Contraindications for surgical closure : VSD + Eisenmenger's syndrome. Because Eisenmenger's syndrome causes irreversible changes in pulmonary artery → No improvement even after surgery.
Surgeries : Dacron patch closure.
Catheter based closure : Occluder device.

Atrial septal defect / ASD 00:30:15

5 to 10% of all CHDs.
Types :

1. Ostium secundum ASD (overall MC ASD).
2. Ostium primum ASD : Associated with mitral valve defects like mitral valve prolapse, mitral regurgitation. Also associated with increased risk of conduction defects.

Rare types of ASD :

- Coronary sinus ASD (unroofed coronary sinus) : Connection between left atrium and coronary sinus.
- Sinus venosus ASD (located at the junction of SVC and right atrium).

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Pathophysiology : Blood flows through atrial septal defect from left atrium to right atrium.

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There is minimal pressure difference between the 2 Atria → No shunt murmur is produced (as there is no turbulence) → Therefore, only flow murmurs.

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Flow murmurs :

- Delayed diastolic murmur.
- Ejection systolic murmur.

Characteristic feature of ASD :
Wide fixed split S2 (second heart sound) seen in both type of ASD.
Normal scenario :
Second heart sound is produced due to closure of aortic and pulmonary valve. On expiration the valves close at the same time and no splitting occurs.
On inspiration the S2 splits as increased venous return to RA → Increases blood flow in RV and pulmonary artery → Delayed closure of pulmonary valve.

ASD : Increased blood flow in right atrium through the ASD → Always delayed closure of pulmonary valve irrespective of inspiration or expiration.

Since both types of ASD show wide fixed split S2 → Echocardiography needs to be done to differentiate the 2 types.
On ECG , ostium secundum ASD shows right axis deviation and Ostium primum ASD shows left axis deviation.
Left axis deviation contributed by left anterior fascicular block and mitral regurgitation.

Complications : Usually start by 2nd /3rd decade.
Least incidence of infective endocarditis (since smooth blood flow won't cause damage to RA).

Active space

Treatment :
Mostly ASD close spontaneously,
So if ASD <8 mm + asymptomatic, observe.

Leave Feedback

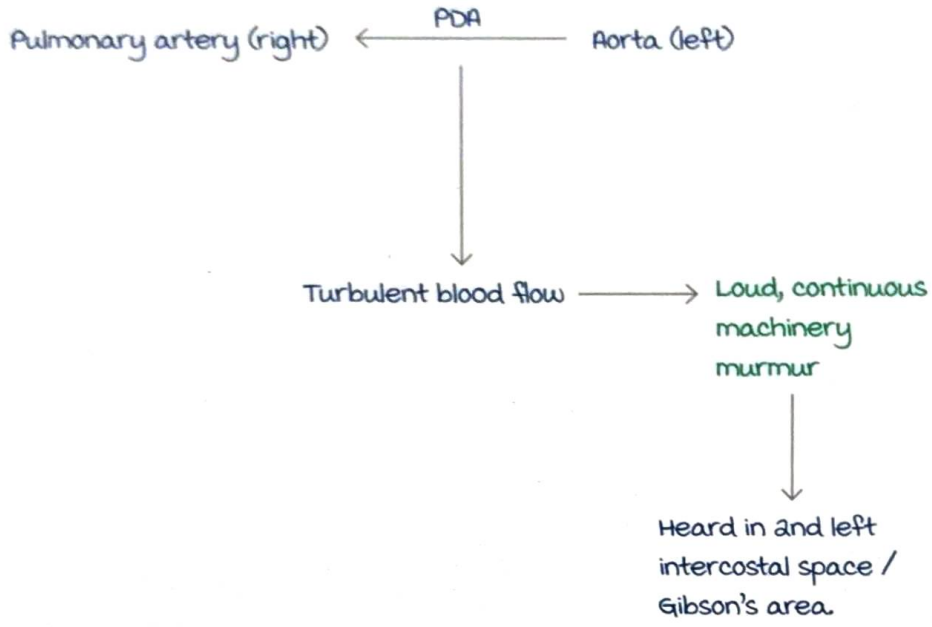
If no spontaneous closure → Surgical closure (done before school entry /<4yrs age) → Patch closure/ catheter based closure with occluder device.

Other indications :

- CCF.
- Size > 8 mm.
- $Q_p : Q_s > 2 : 1$.

Patent ductus arteriosus / PDA

00:45:23



Common in female and preterm babies.

Associated infection : Rubella.

Features :

- Loud, continuous murmur/machinery murmur.
  Other conditions with continuous murmur :
  1. Rupture sinus of Valsalva (RSOV).
  2. Aortico pulmonary window.
  3. AV malformations.
  4. Collaterals (coarctation of Aorta).

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Complications :

- Complications similar to other conditions like CCF, Eisenmenger syndrome etc.
- PDA + Eisenmenger syndrome : The shunting PDA will be reversed due to Eisenmenger syndrome causing differential cyanosis (Upper limbs : Pink, Lower limbs : Cyanosed).

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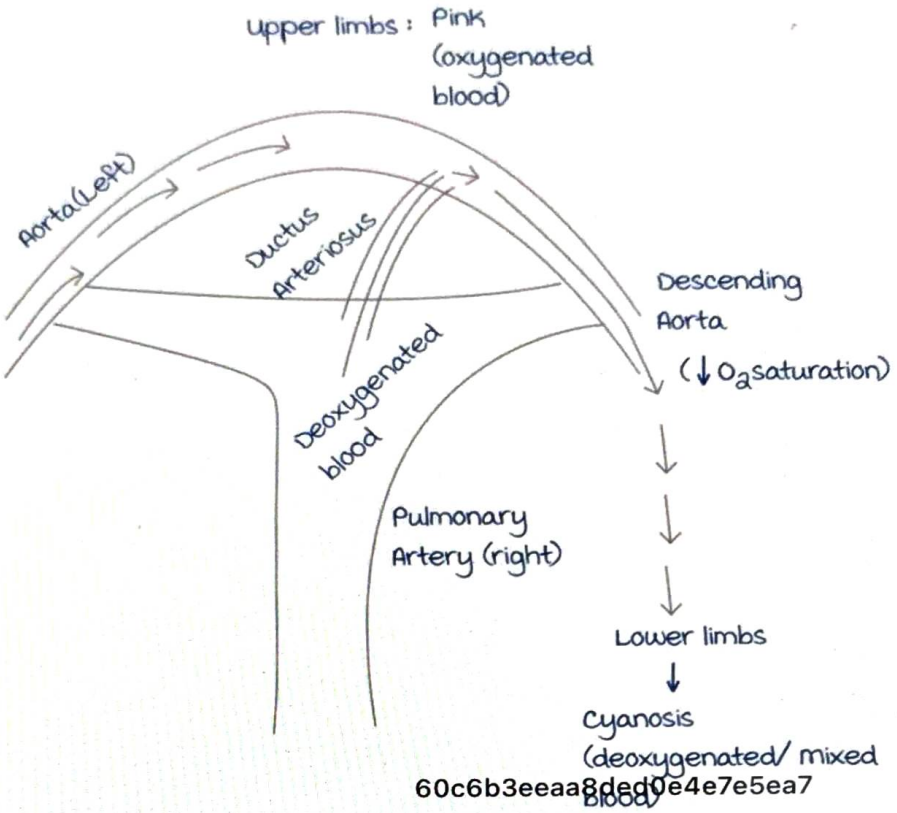
Treatment :

Medical : To Control CCF : Digoxin, Furosemide.

To treat infections : Broad spectrum antibiotics.



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PDA + Eisenmenger's Syndrome

Medical closure of PDA :

Suitable for preterm >> term babies.

It should be started within 2 weeks after birth.

Drugs : PG inhibitors , NSAIDS like Indomethacin & Ibuprofen.

Both drugs are equally effective in closure of PDA. Success rate : 70-80%.

Ibuprofen is the superior drug due to low range of side effects especially Necrotising Enterocolitis/ NEC.

If child is on total enteral feeds : Oral ibuprofen is preferred.

If child is on total parenteral nutrition/partial enteral feeds : IV Indomethacin is preferred as no IV ibuprofen available in India.

Contraindications for medical closure :

- Platelet count <80,000/cu.mm.
- Serum creatinine 1.8 mg /dl or more.

Active space

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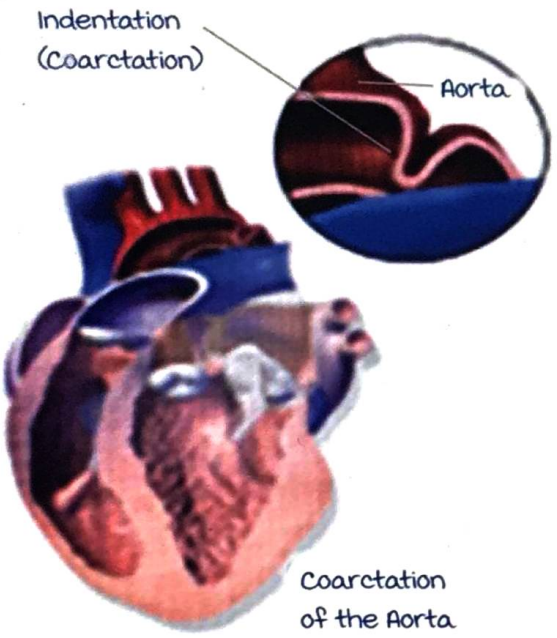
- NEC/feed intolerance.
- Any active bleed.

Surgical closure : If contraindication for medical treatment / failure of medical treatment present (2 doses of Ibuprofen or Indomethacin given and still no closure).
most of term babies usually need surgical closure as medical closure is ineffective.
Ligation / catheter : Occluder device/coil closure can be used.

Coarctation of aorta 01:00:32



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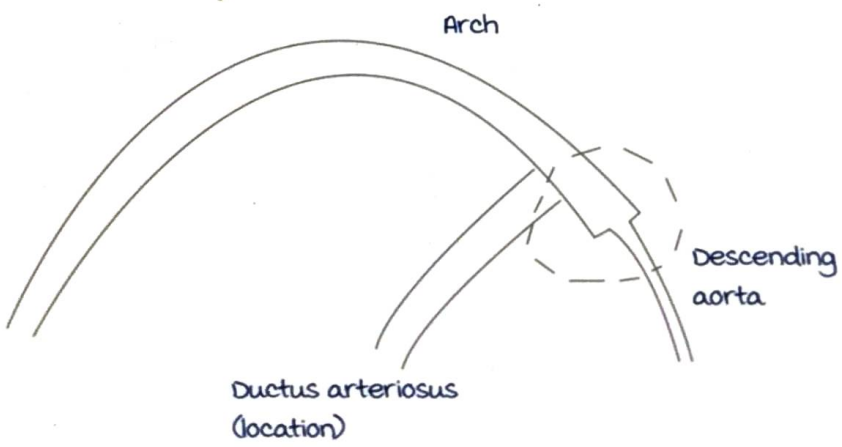

Location of coarctation of Aorta

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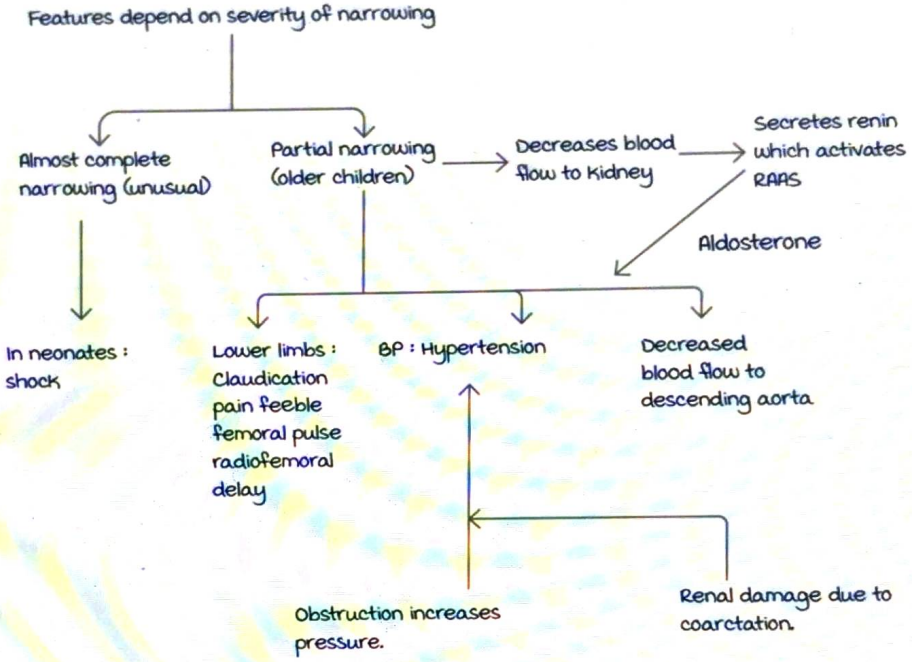
Obstructive lesion / narrowing of aorta.

Location : Juxta ductal / at junction of arch and descending aorta.

Features :

- Lower limbs :
  1. Intermittent claudication pain.
  2. Feeble femoral pulse.
  3. Radio femoral delay.
- BP : Hypertension due to :
  1. Obstruction increasing pressure.
  2. Renal damage due to coarctation.



- Decreased blood flow to descending aorta → Compensation.

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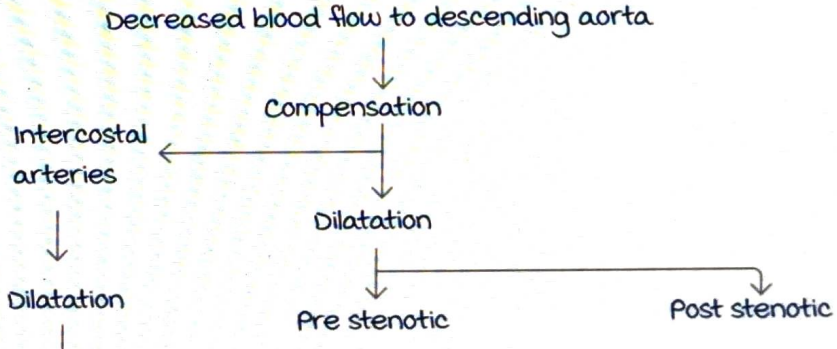

Xray : Inferior Rib Notching on lower border of ribs due to compression from dilated intercostal arteries.

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On X rays : Inferior rib notching and 3 sign seen (prestenotic & post stenotic dilatation) seen.

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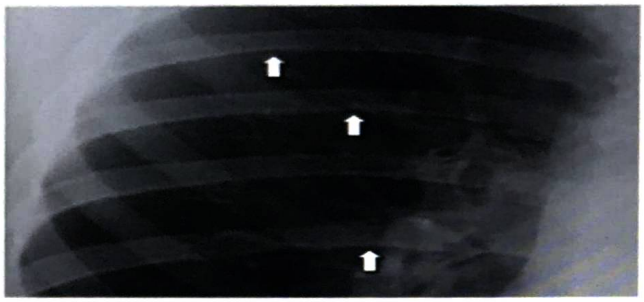
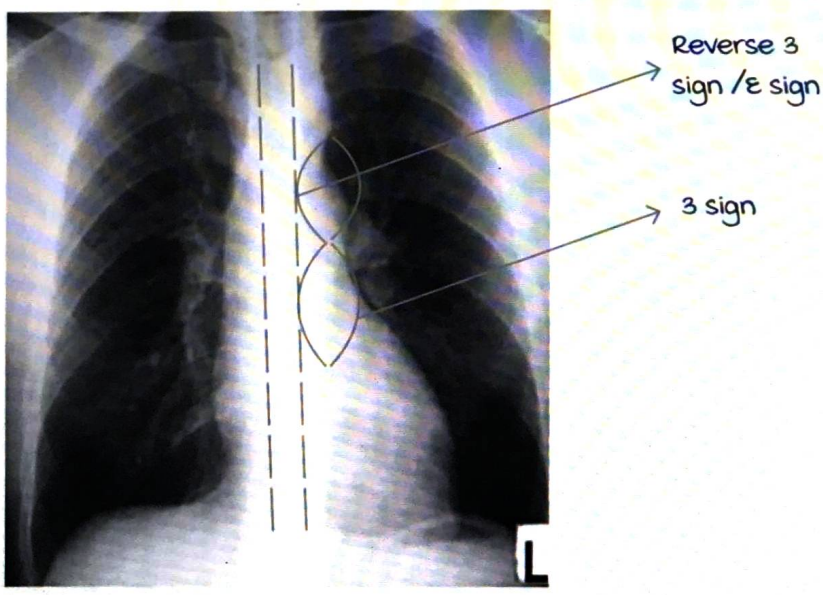


Inferior rib notching in a patient with coarctation of the aorta

On barium swallow, the dilatation will compress the esophagus appearing as a ε sign/reverse 3 sign.



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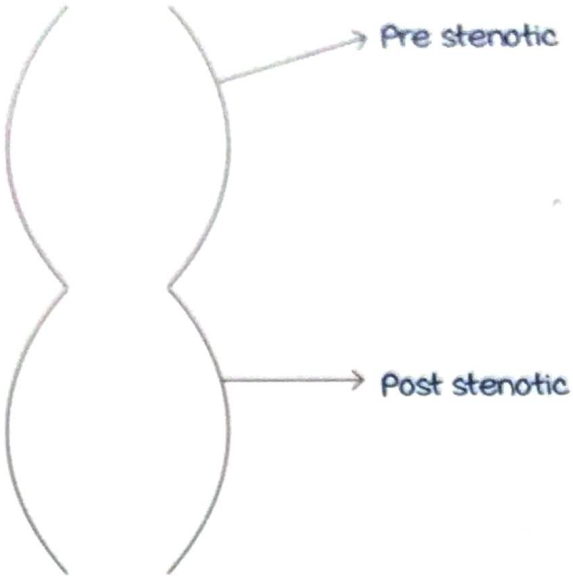

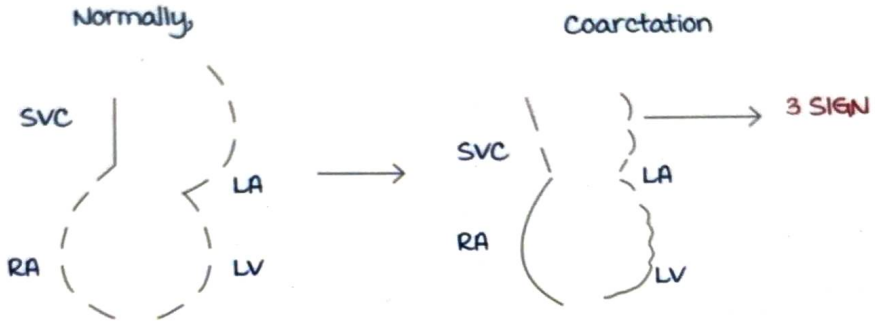

Compensation due to dilatation

Treatment : Angioplasty → Balloon dilatation +/- stenting.

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Active space

CYANOTIC CONGENITAL HEART DEFECTS

Types of cyanotic congenital heart defects :

- Obstructive lesion.
- Mixing lesion (increases pulmonary blood flow).

Obstructive lesions > Mixing lesions.

Obstructive lesions :
Obstruction to the pulmonary blood flow → Decreased pulmonary blood flow → Decreased oxygenation by lung → Cyanosis.

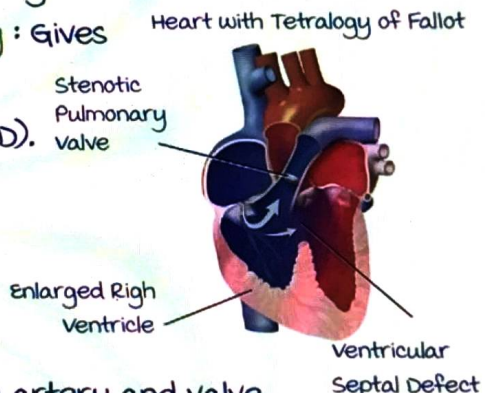
1. Tetralogy of Fallot (overall most common cyanotic heart disease).
2. Tricuspid atresia.
3. Ebstein's anomaly.

Tetralogy of Fallot

00:02:09

Four components of Tetralogy of Fallot (TOF) :

- Overriding of aorta : The aorta after arising from the left ventricle slightly shifts to the right side.
- Right Ventricular Hypertrophy : Gives rise to right to left shunt.
- Ventricular Septal Defect (VSD).
- Sub Pulmonary Stenosis >> Pulmonary stenosis.



Sub Pulmonary Stenosis :

- Stenosis below the pulmonary artery and valve, towards the infundibulum part of the right ventricle.
- Also called right ventricular outflow tract (RVOT).
- Infundibular area hypertrophied → Thick/narrow → RVOT obstruction (main determinant of cyanosis).
- Due to decreased pulmonary blood flow by the RVOT obstruction, the child develops cyanosis.
  The Right to Left shunt through VSD → mixing of oxygenated blood & deoxygenated blood → Cyanosis.

Pentalogy of Fallot : TOF + ASD (Atrial Septal Defect).

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Trilogy of Fallot :

- ASD.
- Pulmonary Stenosis with intact ventricular septum.
- Right Ventricular Hypertrophy.

Consequences of TOF :

- Cyanosis :
  Age is variable as it depends on severity of RVOT obstruction. most of the children develop cyanosis within 4 months after birth.
  Progressive in nature (as the child develops, narrowing of the RVOT also increases).
- Failure to thrive.
- Single second heart sound :
  Only Aortic component of the heart sound is heard. pulmonary component is not heard because of narrowing in the pulmonary region.
- Pulmonary area – Ejection systolic murmur :
  Due to blood flowing through the narrow pulmonary area.
- VSD murmur not heard :
  Large and non-restrictive VSD → No turbulence.

Complications of TOF :

- Infective Endocarditis : Turbulence → Damage to the right ventricle and predisposing to infections.
- Polycythemia : Long standing cyanosis → Chronic hypoxia → Release of erythropoietin → ↑ RBC production.
- Thrombosis : Increased RBCs in blood → Increased viscosity of the blood → Thrombosis/thromboembolism.
- Paradoxical emboli : An embolus from the venous system enters the arterial system via the VSD in a patient of TOF.
- Brain abscess : If a septic paradoxical embolus → Enters CNS → Brain abscess.
- Location of brain abscess in TOF : Parietal lobe.
- Excessive crying, feeding, exertion can cause spasm of infundibulum lead to Tet spells/ hyper cyanotic spells/ hypoxemic spells.

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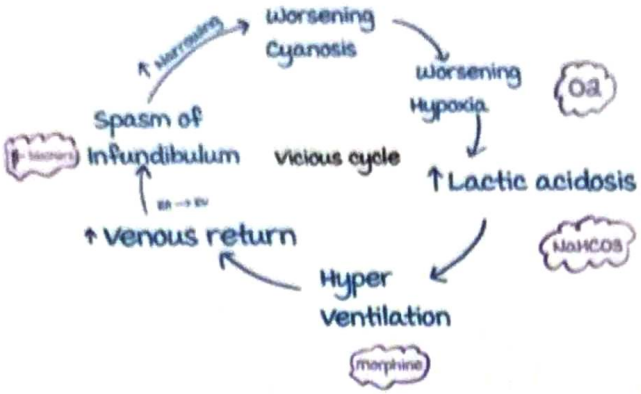
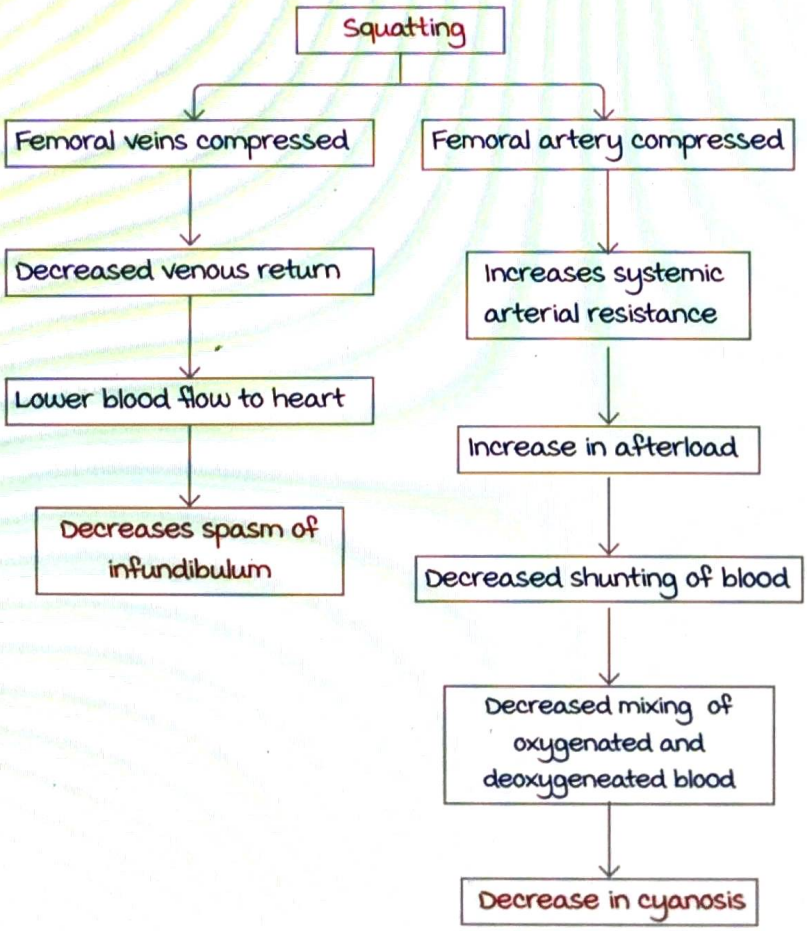
Active space

Vicious cycle of hyper cyanotic spell :



The managing of cyanotic spell is by treating each component of the cycle.

- Hypoxia : Oxygen supply is given.
- Lactic Acidosis : Sodium bicarbonate is given.
- Hyperventilation : Morphine decreases the activity of the respiratory center of the brain.
- To decrease the spasm of the infundibulum : Beta Blockers like Propranalol, Esmolol may be used.
- Children during cyanotic spell, adapt squatting position.



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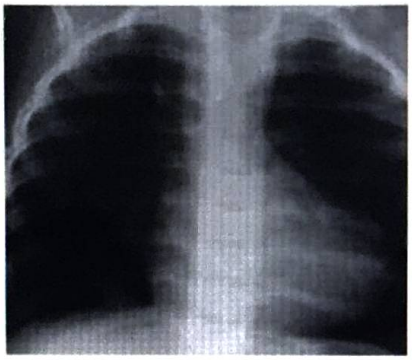
Squatting position is done by older children.
For infants, the mother is advised to bring the baby to a knee chest position during a cyanotic spell.

- α agonistic drugs like Phenylephrine can also be given to increase the systemic arterial resistance.

X-Ray : Boot Shaped Heart or Coer-En-Sabot(due to right ventricular hypertrophy pushing up the apex/left ventricle superiorly).



Coer En Sabot

Treatment :

Definitive treatment :

Early diagnosis is key.
surgery is best done within 3 months after birth (as early as possible).

Palliative treatment :

Shunt surgeries (improve pulmonary circulation).
Various shunt surgeries are :

1. Classical Blalock-Taussig shunt :
   Subclavian artery → Pulmonary artery.
2. Gortex/ modified Blalock-Taussig shunt :
   Subclavian artery is connected to the pulmonary artery via conduit.
   most commonly performed shunt surgery.
3. Waterston shunt :
   Ascending aorta → Pulmonary artery.
4. Potts shunt :
   Descending aorta → Pulmonary artery.

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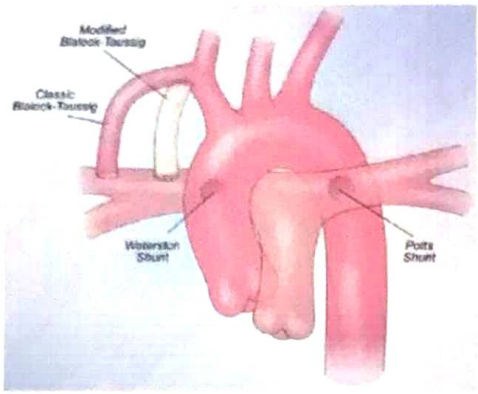

Tricuspid Atresia

00:32:09

Atresia of the tricuspid valve → Small sized RV → Decreased pulmonary blood flow → Cyanosis.

Associated Defects with Tricuspid Atresia :

- Patent foramen ovale or ASD.
- Mixing of blood from right to left side of heart via the ASD → Low O_2 saturation → Cyanosis.
- Ventricular septal defect.

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Tricuspid atresia

Features :

- Cyanosis develops at the time of birth or neonatal period.
- The blood shunts from right to left atrium and ventricle → Overloading of left heart chambers → Congestion on the left side.

ECG : Left axis deviation (important distinguishing feature from other cyanotic HD).

Treatment :

- Should be started as early as possible.
- PGE_1 (Alprostadil) infusion : Keeps the ductus patent (allows blood from aorta to flow into the pulmonary artery).

Active space

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Definitive Treatment :

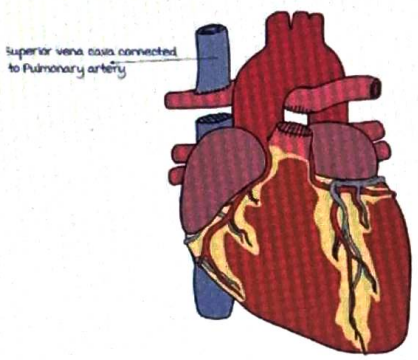
Series of Surgery : Staged shunting procedures.

Modified Blalock Taussig Shunt → Glenn Shunt → Fontan's Shunt.

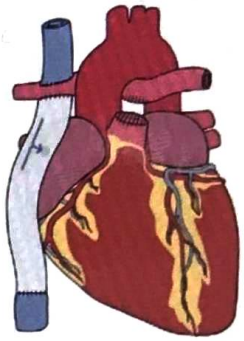
1. Modified Blalock–Taussig Shunt : To improve the pulmonary blood flow.

2. Glenn Shunt : SVC is directly connected to the pulmonary artery → Prevents overloading of left heart chambers and also Increases pulmonary blood flow.

3. Fontan's Shunt : IVC is shunted/connected directly to the pulmonary artery.



Glenn shunt



Fontan's shunt

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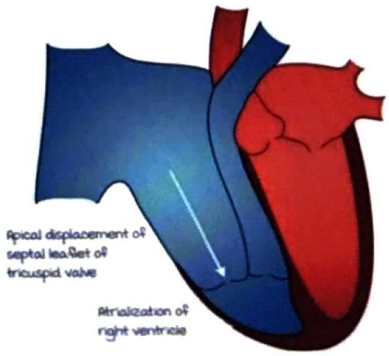
Ebstein's Anomaly

00:42:15

Associated with lithium exposure during pregnancy.

Downward displacement/prolapse of the tricuspid valve → Part of right ventricular volume now becomes part of atrium.

This is known as atrialization of the right ventricle.



Ebstein's Anomaly

Active space

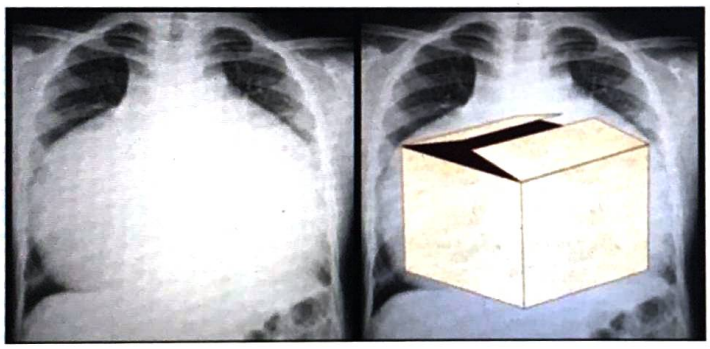
Consequences of Ebstein's Anomaly :

- Tricuspid Regurgitation : Prolapsed valve → Blood from the right ventricle regurgitates to the right atrium.
- RVOT Obstruction : Prolapsed valve partly occludes the blood flowing towards the pulmonary artery → Cyanosis.

Ebstein's Anomaly features :

- Cyanosis at birth or neonatal period.
- Pansystolic Murmur : Due to tricuspid regurgitation.
- Quadruple Rhythm : Irregular blood flow in the right heart, RVOT obstruction and tricuspid regurgitation → Multiple added sounds and ejection clicks are heard.

X-Ray finding in Ebstein's Anomaly :
Due to regurgitation from right ventricle and atrialization of right ventricle, the RA is enlarged.
The RA forms the right heart border which displaces laterally & appears on X-ray as though a box can fit in : Box shaped heart.



Ebstein's anomaly associations :

- Antenatal exposure to lithium.
- Wolf Parkinson White (WPW) Syndrome : Bundle of Kent (accessory pathway connecting the atrium to ventricle) in WPW Syndrome → pre-excitation of the ventricle → predisposing to arrythmias (m/c : Paroxysmal Supraventricular tachycardia).

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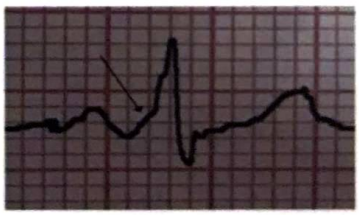
ECG findings in WPW syndrome :
1. Wide QRS complex : >0.08 sec.
2. Short PR interval.



Active space

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3. Delta wave.

Treatment for Ebstein's Anomaly :

- PGE_1 infusion.
- Surgery : Cone repair of the tricuspid valve.

Truncus arteriosus 00:54:38

- Truncus Arteriosus (TA).
- Transposition of Great Arteries (TGA).
- Hypoplastic Left Heart Syndrome (HLHS).
- Total Anomalous Pulmonary Venous Connection (TAPVC).

Mixing of deoxygenated blood with oxygenated blood → Decreased oxygen saturation in the blood → Cyanosis & increased pulmonary blood flow.

Truncus arteriosus 00:55:38

Single arterial trunk into which both the ventricles drain instead of 2 arteries arising from the LV and RV → Deoxygenated & oxygenated blood to mix → Predisposes to cyanosis.

Cyanosis at birth or neonatal period is seen.

Associations : DiGeorge Syndrome (Conotruncal defects are also seen in this syndrome).

Conotruncal defects include :

- Truncus Arteriosus.
- Interrupted Aortic Arch.
- TOF.

Treatment : Rastelli's Procedure.

Hypoplastic Left Heart Syndrome 00:58:21

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Basic Defect : Atresia of Mitral Valve → Hypoplastic left ventricle → Decreased blood flow into the aorta → Small aorta → Hypoplastic Aorta.

Most patients have ASD (which causes mixing of blood).

Decreased flow in the aorta → Systemic circulation gets compromised → Receives very less blood → Shock.

Active space

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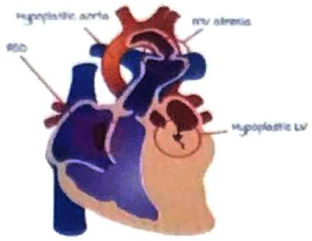
Due to mitral valve block → Congestion of other chambers → Congestive cardiac failure(seen in the neonatal period as well).



m/C cause of CCF in the first week of life : Hypoplastic left heart syndrome.

Treatment :
Initially medical treatment is given : PGE_1 infusion.
This causes improved circulation in the aorta from the pulmonary artery.
Surgery : Norwood's procedure.

Transposition of great arteries/TGA 01:02:26

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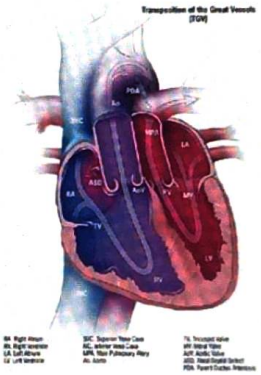
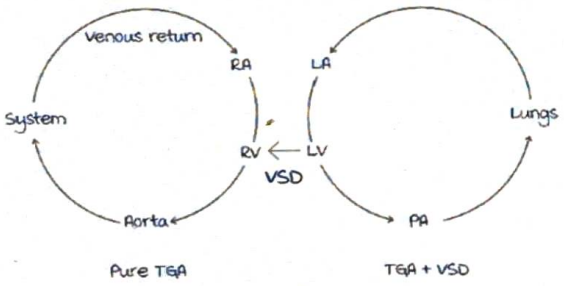
Change in the position of the normal aorta and Pulmonary artery. i.e. aorta arises from the right ventricle and the pulmonary artery arises from the left ventricle.

Parallel circulation is seen in TGA, i.e. there is no cross over between the 2 circulations.





TGA is divided into 2 categories :

- Pure TGA/ TGA with intact ventricular septum : Intense cyanosis at birth or soon after birth.
- TGA with VSD :
    - Pansystolic murmur.
    - Less severe cyanosis (Aorta receives some amount of oxygenated blood from VSD).

Active space

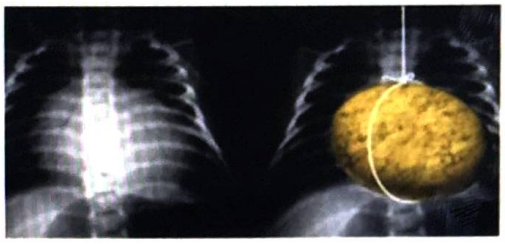
Leave Feedba

- CCF : Due to increased blood entering the pulmonary artery and left heart.

On X-ray : Heart shadow with relatively narrow area above the heart chambers.
Instead of the pulmonary artery and aorta being positioned side to side, they appear positioned on top of each other in A-P view.

This appears narrower than normal on X-ray, like a string attached to an egg.

Egg on string or Egg on side appearance.



Treatment :

Pure TGA : kumarankitindia1@gmail.com

Emergency intervention - neonate presents with intense cyanosis.
PGE, Infusion is given.

Surgery :
1. Rashkind procedure : An atrial septostomy is created at the interatrial septum causing shunt from LA to RA, hence decreasing cyanosis.
2. Jatene's/Arterial Switch Procedure (definitive surgery) : Changing the position of the great arteries, done within 2-4 weeks after birth .

TGA with VSD :
Treatment can be delayed compared to pure TGA (within 3 months after birth).
Jatene's arterial switch procedure can be performed directly.

Active space

Leave Feedback

Total anomalous pulmonary venous connection/TAPVC

01:12:40

Instead of delivering blood into the left atrium, the pulmonary vein drains into the right atrium.

4 types of TAPVC :

- Supracardiac type (m/c).
- Cardiac Type.
- Infracardiac Type.
- Mixed Variety.

Supra cardiac type :

Pulmonary veins join → Vertical vein → Innominate vein (brachiocephalic vein) → Connects to the SVC → Right atrium. Therefore, the oxygenated blood reaches the right atrium instead of the left atrium.

TAPVC patients have patent foramen ovale or ASD → Oxygenated blood reaching the left side of heart leaves via aorta. i.e. mixed blood from the RA via the VSD is supplied by the aorta.

- The blood supplied by aorta is majorly deoxygenated blood causing cyanosis (mild cyanosis).
- In TAPVC pulmonary artery receives more blood → Congestion in lungs and heart → CCF is also seen.
- Due to ASD, wide fixed split of S2 is heard.

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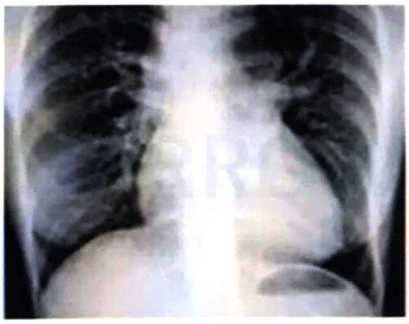
Investigation :

Estimation of O_a saturation in different chambers of heart :

All 4 chambers have the same O_a saturation, since RA supplies all the chambers of heart (RV & LA/LV via ASD).

On X-Ray :

Figure of 8 appearance/ snowman appearance/ cottage loaf appearance (Dilatation of the SVC).



Active space

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Cardiac type :

The pulmonary veins directly connect towards the RA.

Similar features to supracardiac type :

- Mild cyanosis.
- Congestive cardiac failure.
- ASD.

Figure of 8 appearance is not seen in this condition (SVC is not involved).

Infra cardiac TAPVC or Obstructive TAPVC :

Pulmonary veins connect to the IVC or hepatic vein or portal vein.

At the point where the pulmonary veins join the IVC, the pulmonary veins undergo obstructive changes → Decreased draining of blood from pulmonary vein → Pulmonary venous hypertension → Congestion in lungs → Intense cyanosis at birth.

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X-Ray : White out lungs/Ground glass appearance of lungs (pulmonary congestion).

Ground glass appearance of lungs is seen in :

- Respiratory Distress Syndrome.
- Pulmonary Alveolar Proteinosis.
- Obstructive type of TAPVC.

Treatment of TAPVC :

Obstructive Lesion :

- All infra cardiac TAPVCs.
- Surgery done as early as possible.

Non Obstructive Lesion :

- Supra cardiac or cardiac.
- Surgery done in first few months of life.

PGE_1 infusion is contraindicated in infra-cardiac type of TAPVC because blood flows from aorta into pulmonary artery, increasing pulmonary congestion.

Active space

ACUTE RHEUMATIC FEVER

Acute rheumatic fever (ARF) 00:00:14

Post Streptococcal disorder : Group A beta hemolytic streptococci.

Age group affected : School going children (5–15 years).

Incidence : Equal in both males and females.

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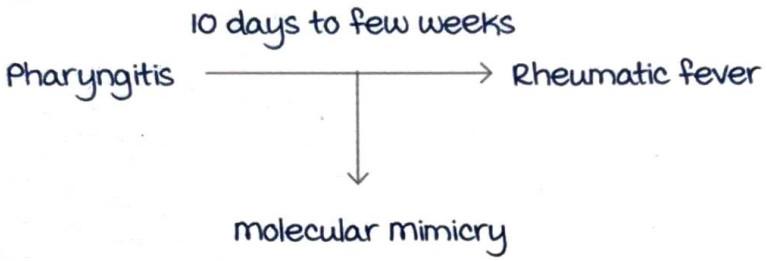
Pathogenesis :



- Following Streptococcal infection, there will be presence of Streptococcal antigens in the body.
- The body produces anti streptococcal antibodies against the antigens and target them.
- Certain body tissues like heart, joint and skin show similarity to these antigens.
- The antibodies cross react with body's own tissues resulting in rheumatic fever →molecular mimicry.

Probability : Normal : 0.3%.

In epidemic : 1–3%.

Clinical manifestations of rheumatic fever 00:04:35

Modified Jones criteria (2015) :

Divided regions of world into two :

- Low risk population.
- Moderate to high-risk population (India) :
  ARF (Acute rheumatic fever) incidence > 2/1 lakh school going children.
  RHD(Rheumatic heart disease) prevalence > 1/1000 population.

Active space

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Criteria includes :

Major criteria (moderate to high risk population) :

1. Pancarditis :

   Either clinical or sub clinical (ECHO findings suggestive of carditis).

2. Arthritis :

   mono/polyarthritis/polyarthralgia (In low-risk population, only polyarthritis is considered a part of major criteria).

3. Sub cutaneous nodules.
4. Erythema marginatum.
5. Sydenham's chorea.

Minor criteria :

Moderate to high risk population	Low risk population
Monoarthralgia.	Polyarthralgia
Fever ≥38 degree.	Fever ≥38.5 degree.
ESR ≥30 mm/hour and/or CRP ≥ 3 mg/dl.	ESR ≥ 60 mm/hour and/or CRP ≥ 3 mg/dl.
Prolonged PR interval.	

Essential criteria :

Evidence of preceding streptococcal infection :

1. Elevated ASO titer (Anti streptolysin O titer) :
   Preferred investigation.
2. Throat swab culture (not usually preferred).
3. History of preceding sore throat (>50%).

Diagnosis :

First episode : 2 major criteria OR

1 major + 2 minor criteria.

Recurrence

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2 major criteria OR

1 major+ 2 minor criteria OR

3 minor criteria.

In addition to these, essential criteria should also be fulfilled to confirm the diagnosis.

Active space

Leave Feedback

Carditis 00:16:42

Pancarditis : All 3 layers affected.
Most common feature of ARF (90%).
Early finding of rheumatic fever (within 2 weeks).
Most serious : Permanent damage of valves can occur.
Diagnosis :
Clinical → pericarditis : Chest pain + frictional rub.

Valve involvement :
Most common valve : Mitral valve > aortic valve.
Mitral valve involvement : Common in females. Aortic more common in males.
Result in

1. Mitral valvulitis : Carey coombs murmur (delayed diastolic murmur).
2. Mitral regurgitation :
   - Soft S_1.
   - Pan systolic murmur.
   - Cause hemodynamic overload and precipitate Left ventricular failure.

Main reason for morbidity/ mortality in ARF : Left ventricular failure.

Arthritis 00:22:34

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Large joints affected (knee, ankle, elbow).
Migratory arthritis.
Early finding.
Found in 30–50% of affected children.
Excellent/dramatic response to aspirin.
No residual joint damage → Complete recovery.

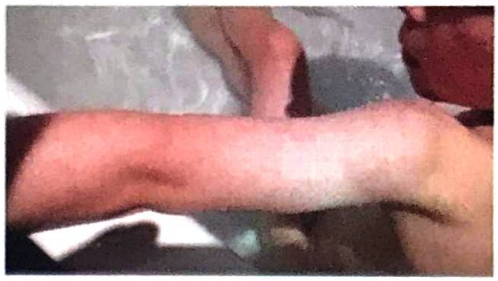
Erythema marginatum 00:24:44

Rare finding.
If present, found in early stages.
Erythematous lesion with pale center.
Over time the rashes coalesce with each other forming serpinginous rash.

Active space

Leave Feedba

- Non pruritic.
- Starts in trunk an limbs.
- Not found in face.
- Induced by warmth heat.



Erythema marginatum

Sub cutaneous nodules 00:26:51

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Late finding (6 weeks after onset).

Painless nodules.

Present around bony prominences (elbow, shin, occiput, ankle), where tendons attach to bone.

Presence of nodules is associated with high risk of carditis.

Sydenham's chorea 00:28:15

Last manifestation (3 months after onset).

Semi purposeful, jerky movements of limbs resulting in abnormal coordination and abnormal posturing.

Associated manifestations :

- Hypotonia.
- Emotional lability.
- Jerky speech.

Signs :

- Milkmaid grip sign : Alternate tightening and loosening of grip.
- Darting tongue sign : Unable to keep the tongue protruded out for examination.

Self resolving (2-6 weeks).

If not resolving, treat with diazepam, haloperidol.

Management 00:31:30

Treatment :

Strict bed rest for 2 weeks.

Penicillin :

Inj. Penicillin G (Benzathine) single dose/ Oral Penicillin V 250 mg 4 times /day x 10 days.

Active space

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Allergic to Penicillin : Erythromycin 250mg 4 times a day x 10 days.

Anti inflammatory medications :

Steroids :

Preferred (especially in cardiac problems).

Oral Prednisolone (2mg/kg/day to a maximum of 60 mg/day) For 3 weeks. Taper over 9 weeks.

Aspirin :

90-120mg/kg/day in 4 divided doses x 10 weeks.

Taper over next 2 weeks.

Total duration of treatment with anti-inflammatory medications in ARF : 12 weeks.

Secondary Prophylaxis :

Prophylaxis given to prevent further episodes of ARF and cardiac issues.

- Penicillin (Benzathine/Penicillin G) :
  > 30 kg : 12 lakh IU every 3 weeks.
  < 30 kg : 6 lakh IU every 2 weeks.
- Oral Penicillin V : 250 mg twice a day.
- Allergic to Penicillin : Erythromycin 250 mg twice a day.

Duration of prophylaxis :

Depends on problems encountered in initial episode of rheumatic fever.

- No carditis : Prophylaxis for next 5 years or till 18 years of age, whichever is longer.
- Carditis (without RHD) : Next 10 years or till 25 years of age, whichever is longer.
- Established RHD/undergone surgery :
  Till the age of 40 years.
  Ideally given life long.

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CONGENITAL ANOMALIES OF KIDNEY & URINARY TRACT

Anomalies of renal system

00:00:16

1. Renal parenchymal anomalies.
   - Upper part of renal tract.
2. Distal urinary tract anomalies.
   - Urinary bladder, urethra etc.

Renal parenchymal anomalies :

1. Renal agenesis (absence of development of Kidney).
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2. Dysplasia (presence of primitive ductal structures in Kidney due to abnormal metanephric differentiation).
3. Fusion defects (Horse-shoe Kidneys).

Dysplatic disorders :

Cystic dysplasia (unilateral >> bilateral).

1. Unilateral renal dysplasia/Multi-cystic Dysplastic Kidney (MCDK):
   - more common in the left side.
   - multiple cysts may replace normal tissue.
   - Renal function on affected side absent.
   - Contralateral Kidney → Hypertrophy.
   - Overall, renal function normal (GFR/ S.creatinine normal).
   - Cysts → Spontaneous involution/regression by 5-7 years of age (function won't be normal).
   - Features :
     1. Abdominal mass (loin region).
     2. Otherwise, asymptomatic.

m/c cause of palpable abdominal mass in newborn.

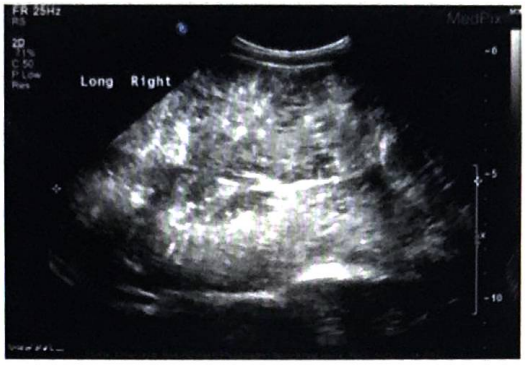
m/c cause of cystic disorder of Kidney.

Active space

ARPKD 00:07:33

Autosomal recessive polycystic kidney disease bilateral

- Defect in PKHD 1 gene (short arm of chromosome 6) → Fibrocystin protein → Fibrosis + cyst formation in kidneys.
- Liver also affected.
- Features :
  1. Bilateral abdominal mass (loin area).
  2. Overall renal function decreased → End stage renal disease (ESRD).
  3. Compress renal blood vessels → Ischemia in kidney → Renin release → Activates RAAS (Renin-Angiotensin-Aldosterone system) → ↑Aldosterone → Hypertension.
  4. Hepatic fibrosis + cysts → Liver cirrhosis/chronic liver failure.



- Treatment : Dual transplantation (both kidney & liver).
- Ultrasound :
  1. Hyperechoic kidneys.
  2. Absence of cortico-medullary differentiation.

Agenesis of kidney 00:13:46

Severe ARPKD → Dysfunctional kidneys in utero → Potter's syndrome

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Bilateral agenesis of kidney → Potter's syndrome.

Potter's syndrome :

No urine output from fetal kidneys → Severe oligohydramnios → Compression of fetus by amniotic sac → Potter's syndrome.

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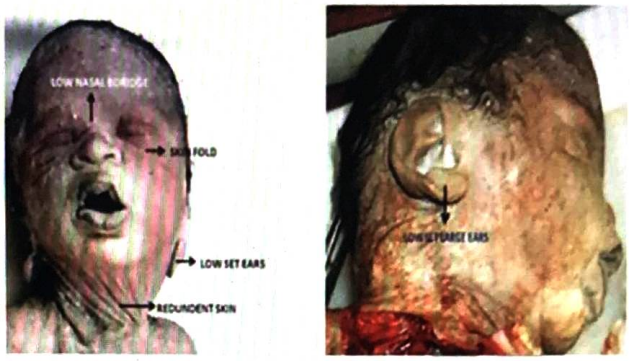
Features :

- Potter's facies :
  Widely separated eyes, broad and flat nose (parrot beak appearance), low set ears, receding (small) chin, lot of skin wrinkles.
- Fetal compression :
  Thorax → Pulmonary hypoplasia → Death immediately after birth.
  Limb abnormalities : CTEV (Congenital talipes equinovarus).



Potter's syndrome

Unilateral renal agenesis 00:19:13

One kidney normal → Hypertrophy → Renal function normal.
Unilateral > bilateral.
Can occur in association with other anomalies
VACTERL :

- Vertebral defects.
- Anorectal malformations.
- Cardiac defects.
- Tracheo Esophageal fistula.
- Renal anomalies.
- Limb anomalies.

Also associated with single umbilical artery.

Renal fusion anomalies 00:21:16

Horse-shoe kidneys :

- Lower poles of kidneys are fused in the midline → Isthmus (fused portion).

Occurs in 1 in 400–500 live births.

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- Associated with Turner's syndrome (30%)
- Complications :
  1. Ureteral obstruction → Hydronephrosis
  2. Renal stasis → Renal stones.
  3. Recurrent UTIs.
  4. Increased risk of Wilm's tumor(4 times riskier).

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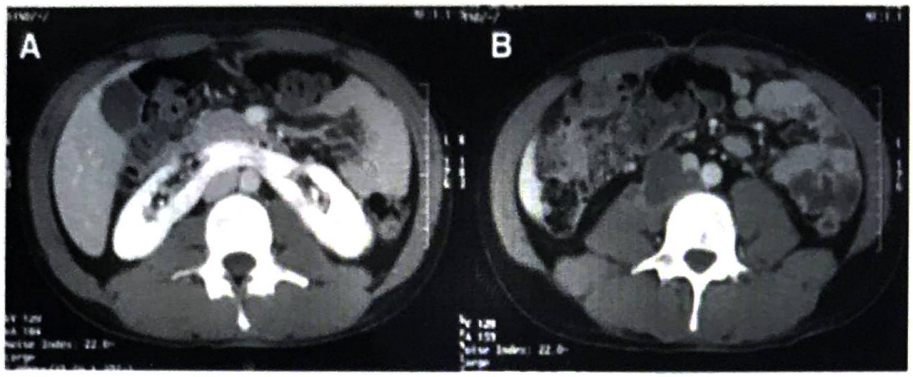

Horse shoe Kidney

Distal urinary tract anomalies 00:25:13

Anomalies outside the kidney (ureters, bladder, and urethra).

Obstructive uropathy :

Etiologies :

1. Ureteropelvic junction obstruction.
2. Posterior urethral valves (PUV).
3. Severe bladder malformation.
4. Eagle-Barrett (Prune Belly) syndrome.

Features :

1. Hydronephrosis → Abdominal mass.
   - Hydronephrosis : 2nd m/c cause of abdominal mass in a new born baby, can also present antenatally.
2. Abdominal pain (loin region).
3. Recurrent UTIs.
4. Poor urine stream.
5. Increased risk of renal stones.

Investigations :

- Antenatal ultrasound : Antenatal (2nd trimester).
- USG KUB
- Micturating cystourethrogram (MCU) : Postnatally

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Ureteropelvic junction (UPJ) obstruction :

- Overall m/c cause.
- Due to abnormalities of smooth muscle/fibrous bands → Narrowing + obstruction.
- Unilateral (left > right).
- Antenatally : Hydronephrosis.
  Postnatal : Abdominal mass (loin region).
- Recurrent episodes of abdominal pain and vomiting (Dietl's crisis) : In old children.
- Management :
  Resolve spontaneously after 48–72 hours post birth
  If persistent, surgical correction : Anderson Heyne's pyeloplasty.

Posterior urethral valves

00:32:50

- Seen in males, m/c severe cause of Obstructive uropathy.
- Severe condition.
- Valves in lower part of prostatic urethra, below/distal to verumontanum, extending till external urethral sphincter, cause urinary obstruction.

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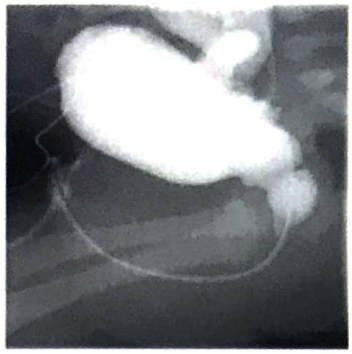
- Area proximal to the obstruction becomes dilated.
- Trabeculations of the bladder wall due to abnormal dilatation of the bladder wall.
- Hydroureteronephrosis.
- Features :
  1. Abdominal mass.
  2. Poor urinary stream → Dribbling of urine during micturition.
  3. Increased risk of urinary tract infections.
- Diagnosis : Micturating cysto urethrogram (MCU)/ voiding cysto urethrogram (VCUG).

Active space

- Management :

1. Insert a feeding tube or feeding catheter to relieve pressure.
   Foley's catheter is not used in children as it may cause spasm.
2. Endoscopic fulguration of the valves (destruction) :
   Definitive treatment.
   Only done if serum creatinine levels are normal.
3. Temporary vesicostomy if serum creatinine is high.



Posterior urethral valves

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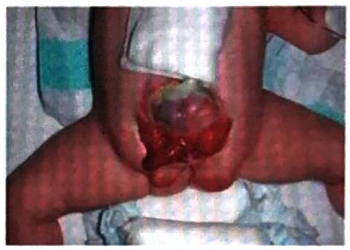
Severe bladder malformation

00:38:54

Ectopia vesicae/bladder exstrophy :
Associated with other malformations :

1. Cryptorchidism or undescended testis.
2. Epispadias.
3. Split pelvis.



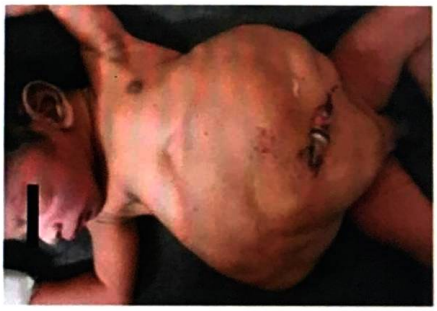
Bladder Exstrophy

Severe form of bladder exstrophy : Ileum + Bladder exposed → Cloacal exstrophy (Elephant trunk ileum).

Eagle-Barrett (Prune belly) syndrome :
Due to:

1. Deficient abdominal wall muscles → Abdominal skin wrinkled.
2. Cryptorchidism.
3. Mega ureter and mega bladder (dilated).

AKA Triad syndrome.



Prune Belly Syndrome

Active space

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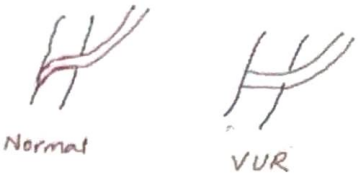
Reflux nephropathy

00:43:17

Vesico-ureteric reflux :

Occurs due to :

- No oblique insertion of bladder.
- Short/small submucosal tunnel.



Micturition → Urine flows back from bladder to ureter.

Can be inherited (Autosomal dominant).

Females > Males.

VUR → Retrograde urinary flow → Recurrent UTIs.

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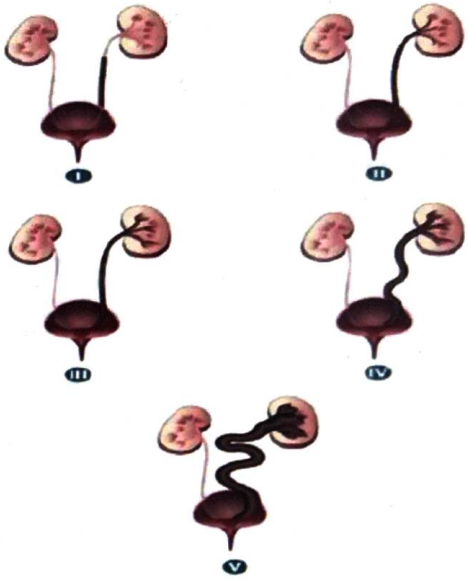
m/C anomaly associated with recurrent UTIs : VUR.

High grade VUR → Renal scarring → Hypertension/ renal insufficiency → Reflux nephropathy.

IOC : MCU/VCUG.



Grades of vesicoureteric reflux

Grading :

Grade 1 : Partial reflux.

Grade 2 : Complete reflux.

Grade 3 : Complete reflux + Dilated ureter.

Grade 4 : Tortuosity of ureter.

Grade 5 : Distortion of renal pelvi-calyceal impression.

Treatment :

- Spontaneous resolution after birth.
- Antibiotics prophylaxis (single time → Bed time dosage) :
  1. Cephalexin < 6 months.
  2. TMP-SMZ (cotrimoxazole) > 6 months.
  3. Nitrofurantoin > 6 months.

  Grade 1/2 → Till 1 year.

  Grades 3-5 → Till 5 years.
- Antibiotic prophylaxis fails → Breakthrough UTI → Surgical correction of VUR (Reimplantation of ureter into bladder in a oblique way to prevent the reflux).

Active space

301
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GLOMERULONEPHRITIS

Glomerulonephritis (GN) 00:00:20

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Hallmark : Hematuria → Gross : Cola coloured urine.
→ microscopic : > 5 RBC/hpf in uncentrifuged.
> 10 RBC/hpf in centrifuged.

Additional findings :

- RBC cast.
- Dysmorphic RBCs > 20 % (Diagnostic for hematuria associated with glomerulonephritis).
- Mild proteinuria.
- Massive proteinuria is seen in nephrotic syndrome.
- Edema : Pedal, periorbital.
- Presence of hypertension : Child will be admitted for BP monitoring and antihypertensive administration.
- Oliguria
- Overall most common condition : Post- Streptococcal glomerulonephritis.
- Organism : Group A beta Haemolytic Streptococcus.
- Infections : Pharyngitis.
  Skin infection (Pyoderma Impetigo)
  Post infection → Lag period → GN.

Lag period : Few weeks post-infection disease develops.

Pathophysiology 00:05:10

At lag period, streptococcal antigen and anti-streptococcal antibodies combine forming immune complex and deposit in kidney.

Type of infection	Strain	Lag period
Respiratory infection (common during winter)	M4, M12 commonly M 1, M 24	1-2 weeks
Skin infection (common during summer)	M 49	3-6 weeks

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Features :

Age group : School going age (5 to 8 years).

Investigations :

- Antibody titre :

60c6b3eeaa8ded0e4e7e5ea7 ASO titre increased.

(ASO → Antitreptolysin O).

If ASO is normal → Anti DNAase B increased.

- Serum C_3 : Decreased as compliment used for immune complex formation.
- Renal biopsy :

Rarely indicated if :

Low C_3 > 12 weeks.

Impaired renal function > (7 – 10) days.

Findings seen in :

Immuno-fluorescence microscopy → Deposit of IgG and C_3 in capillary walls seen as Lumpy-Bumpy/ Starry Sky appearance.

Electron microscope → Sub-epithelial immune complex deposits called Humps.

Management

00:10:50

- Symptomatic treatment.
- Post infectious so will resolve by itself.
- Anti-hypertensive drugs → Amlodipine, Nifedipine.
- Natural disease course is resolution in few days to few weeks.
- General guidelines :

ASO titre and BP will be normal by (4 – 6) weeks.

C_3 → Normal by (8–12) weeks.

microscopic hematuria can persist up to (1 to 2) years.

- Post streptococcal condition :

Rheumtic fever.

PSGN.

- Antibiotic administration can prevent rheumatic fever but not PSGN.

Active space

IgA Nephropathy

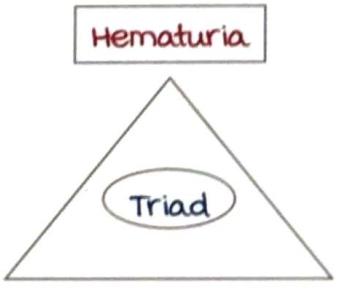
00:14:28

- Primarily because of IgA deposition in Kidney.
- AKA Berger's disease.
- Most common cause of chronic and recurrent nephritis.
- Recurrent episodes follow any respiratory infection.
- Lag period : 1 - 2 days.
- Investigation :
  - C_3 and ASO Titre - Normal.
  - Renal biopsy : Diagnostic.
  - Mesangial proliferation + IgA depositions seen.
- In 10-20 % children with IgA nephropathy can develop chronic Kidney disease in adulthood hence follow-up.
- Management :
  - ACE inhibitor /ARBs → Delays progression of Kidney diseases hence decreasing the occurrence of CKD.
  - Fish oil (rich in PUFA, omega-3): Decreases entry of macrophages into glomerular mesangium which is the initiating factor for IgA deposition.

Hereditary nephritis

00:18:58

- AKA Alport syndrome.
- m/c mode of inheritance → X-linked dominant.
- Also have X-linked recessive and autosomal inheritance.
- Defective gene COL_4A_5 codes for Type 4 Collagen (Alpha chain).
- Triad :



Ear manifestation : High tone / high frequency / SNHL.	Ocular manifestation : On slit lamp examination anterior lenticonus (pathognomic). Others : Corneal erosion, macular flecks.

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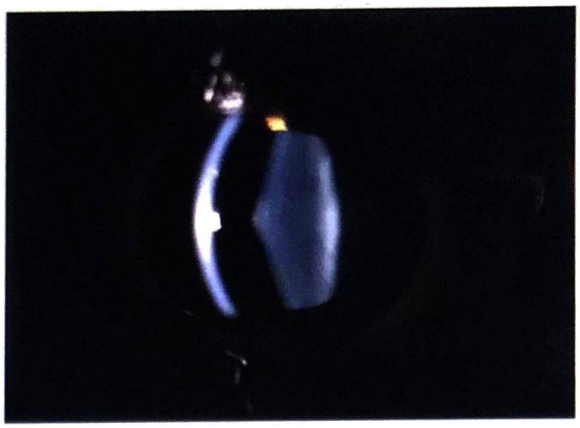

Anterior Lenticonus

Investigation :

- Biopsy shows irregular thickening and thinning of basement membrane + Lamellations present : Basket weave appearance.
- Genetic testing for COL_4A_5 gene defect.

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Treatment :

- Supportive treatment with ACE inhibitors can modify progression or decrease incidence of CKD in children. However, most children develop end-stage kidney disease by 2nd or 3rd decade.
- Renal transplantation (for prolonging survival).

Active space

NEPHROTIC SYNDROME

Definition

00:00:18

Key feature : massive proteinuria → > 40 mg/m^2/hour.

1. Urine Protein : Creatinine ratio > 2.
2. Urine dipstick 3+ /4+.

Done in the first morning sample (spot test).

Other features :

1. Hypoalbuminemia (serum albumin < 3 g/dL).
2. Generalized edema. 60e6b3eaaa8ded0e4e7e5ea7
3. Hyperlipidemia due to increased lipoprotein synthesis in liver (serum cholesterol > 200 mg/dL).

Diagnosis of nephrotic syndrome :

Proteinuria + hypoalbuminemia/edema.

Hyperlipidemia is not part of criteria as per 2021 guidelines.

Disorders associated :

1. Minimal change disease (MCD) :
   - Overall m/c condition associated with nephrotic syndrome in children.
   - Loss/fusion/effacement of foot processes of podocytes → Loss of negative charge → Loss of protein in urine → Proteinuria.

2. Significant change disorders :
   - FSGS (Focal segmental glomerulosclerosis).
   - MN (Membranous nephropathy).
   - MPGN (Membranoproliferative glomerulonephropathy).

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	Minimal change disease	Significant change disease
Age at onset	2–6 years	> 10 years
Gender	Males > females	Males = females
Hematuria	Absent	Present (nephritic onset nephrotic syndrome)
Blood pressure	Normal	May have hypertension (FSGS)
C3	Normal	May be low (MPGN type I)
Selectivity of proteinuria	High selectivity	Low selectivity
Response to steroids	Excellent response	Bad response (unsatisfactory)
Prognosis	Always good	Variable

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Steroids in MCD

00:11:22

MCD shows excellent response to steroids.

Remission : A condition where child responded by reduced protein excretion (< 4 mg/m^2/day) or urine PCR < 0.2 or urine dipstick nil or trace for 3 consecutive early morning spot tests.

Treatment schedule :
Oral prednisolone daily 2 mg/kg or 60 mg/m^2 for 6 weeks.
Maximum dose : 60 mg/day.

Followed by alternate day oral prednisolone 1.5 mg/kg/day or 40 mg/m^2 for 6 weeks.
Maximum dosage : 40 mg/day.

Typical course of nephrotic syndrome characterized by relapses.

1. Massive proteinuria > 40 mg/m^2/hour.
2. Urine Protein : Creatinine ratio > 2
3. Urine dipstick 3+/4+ for 3 consecutive spot tests.

Occurs till 2nd decade of life.

Active space

Treatment :

- Daily steroids till child achieves remission.
- Followed by alternate day steroid for the next 4 weeks.

Congenital nephrotic syndrome 00:17:45

Early onset nephrotic syndrome (< 3 months).
Associated with other genetic disorders :

Gene	NPHS 1	NPHS 2	WT 1
Codes for	Nephrin	Podocin	
Inheritance pattern	Autosomal recessive	Autosomal recessive	Autosomal recessive
Pathology	microcystic dilatation of proximal tubules	FSGS	Diffuse mesangial sclerosis

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WT-1 gene mutation is associated with a syndrome known as Denys Drash syndrome, which is characterised by :

- Ambiguous genitalia in a male child.
- Increased risk of Wilm's tumour.

Important terms in nephrotic syndrome 00:20:49

FRNS (Frequently Relapsing Nephrotic Syndrome) :

- ≥ 2 relapses in 1st 6 months or
- ≥ 3 relapses in any 6 months or
- ≥ 4 relapses in 1 year.

SDNS (Steroid Dependent Nephrotic Syndrome) :

- Develops ≥ 2 consecutive relapses while on alternate day steroid treatment or post 14 days after stopping steroid treatment.

Management :

- Alternate day low dose prednisolone 0.5–0.7 mg/kg.
- Daily in case of infections : 5–7 days.
  (Infections can precipitate relapse).

If patient able to maintain stable remission while on steroid →

Active space

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Decrease dose to 0.2–0.3 mg/kg for 6–12 months and stop.
Despite treatment, relapse developed or threshold becomes
[illegible] sparing agents.

Choice of steroid sparing agents depends on :

1. Threshold > 1 mg/kg.
2. Presence of features of complicated relapse (Relapse with life threatening complications) like :

- Hypovolemia.
- Life threatening infections (peritonitis/cellulitis).
- Thrombosis (increased fibrinogen, low levels of anticoagulant proteins).

m/c cause of peritonitis in nephrotic syndrome : Streptococci pneumoniae.

If any of the above conditions are seen :

- mmF (mycophenolate mofetil).
- Cyclophosphamide.

If none of the above conditions seen :

- Levamisole.
- mmF.

Steroid Resistant Nephrotic Syndrome (SRNS) 00:28:56

No response to steroids even after using daily steroids (2mg/kg/day) for 6 weeks.
Treatment :

- Before starting treatment, do renal biopsy.
- Calcineurin inhibitors (Tacrolimus >> Cyclosporine).

Tacrolimus is contraindicated in seizures & hypoglycemia.

Active space

Live vaccines administration in nephrotic syndrome :

1. If steroids (≥ 2mg/kg/day for ≥ 14 days) used → Avoid live vaccines for 1 month after stopping steroids. (can lead to immunosupression and decreased production of Antibodies.)
2. If cyclophosphamide used → Avoid live vaccines for 3 months after stopping.
3. Other agents → Avoid live vaccines for 1 month.

Recommended vaccines :

1. Pneumococcal vaccine (PCV 10/13, prefered conjugate is 13 valent) given > 6 months age.
   2 doses, 8 weeks apart.
2. Varicella vaccine (live attenuated) given > 15 months age.
   2 dose, 4-8 weeks apart.
3. Influenza vaccine given > 6 months age.
   Annually.

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INHERITED TUBULAR DISORDERS

Common features of inherited tubular disorders :

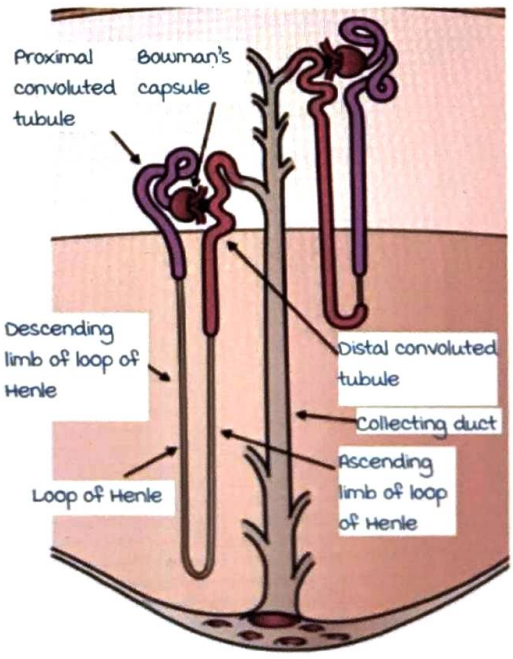
- Polyuria :
  most important function of tubules is to concentrate urine. Dysfunction of tubules can lead to excess urine output from the kidney.
  Polyuria is defined as daily urine output >2L/m^2 body surface area.
- Polydipsia : Excessive thirst.
- Failure to thrive : Due to impaired absorption of glucose and nutrients from the tubule.

Disorders	Site of defect	Defect
Bartter's syndrome	Ascending limb of loop of Henle	Na^+ K^+ $2Cl^-$ co-transporter Mimics Furosemide.
Gitelman syndrome	Distal convoluted tubule (DCT)	Na^+-Cl^- cotransporter. Mimics Thiazides.
Liddle syndrome	Collecting duct.	Gain of function mutation in gene that codes for ENaC (Epithelial Sodium channel) ⟶ Increased activity of the channel.

Inheritance : kumarankitindia1@gmail.com

Autosomal recessive : Bartter and Gitelman Syndromes.

Autosomal dominant : Liddle Syndrome.



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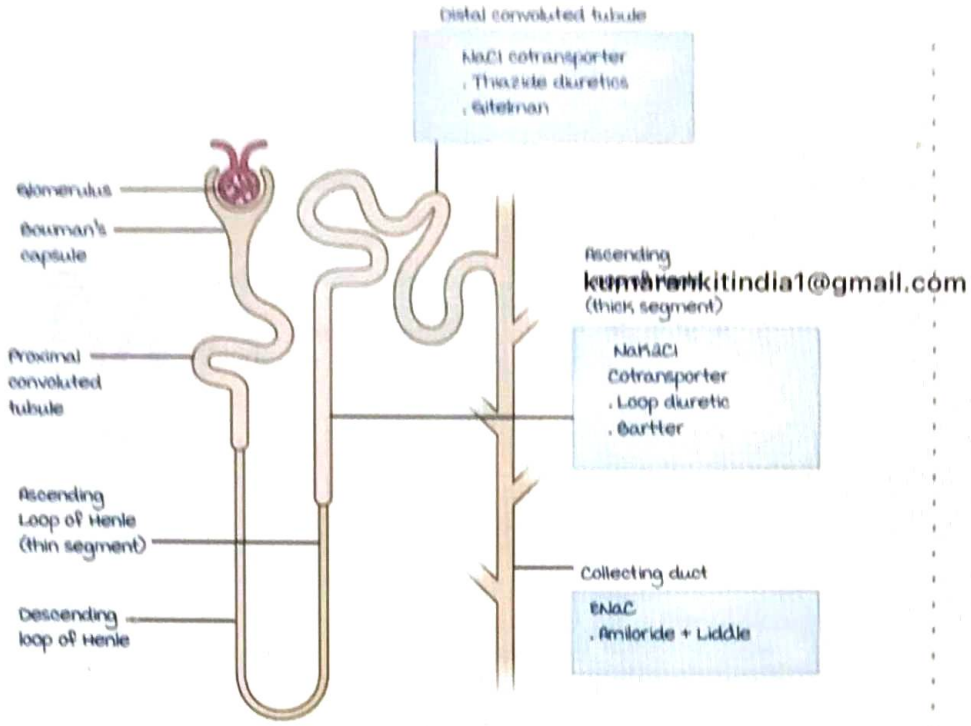

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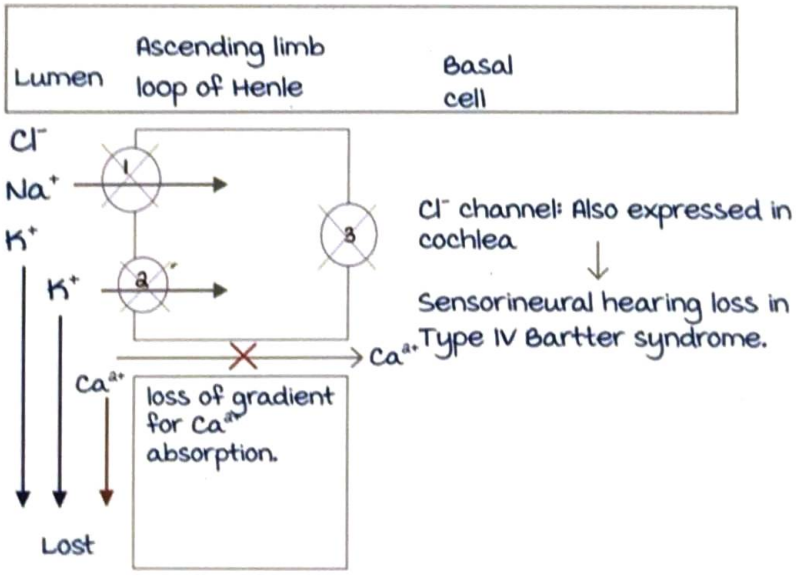
Bartter syndrome

00:05:35

Inheritance : Autosomal recessive disorder

Onset : Infancy.

Loss of function :



1. Na^+-K^+-$2Cl^-$ co-transporter.
2. Renal outer membrane Potassium (ROMK) channel.
3. Cl^- channel (Gene : Bartin)

Active space

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Consequences :

1. Loss of Na^+, K^+, Cl^- in urine.

↓

Dehydration/ ↓intravascular volume.

↓

Stimulates Renin release from Kidney.

↓

Activates Renin – angiotensin – Aldosterone mechanism.

↓

- Na^+ absorption in collecting duct
- K^+ secretion → K^+ lost in urine

↓

Hypokalemic hypochloremic metabolic alkalosis.

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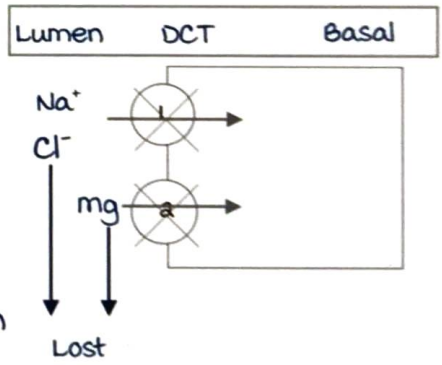
2. Loss of Ca^{2+} in urine → Hypercalciuria → Stones

Gitelman syndrome

00:12:03

Onset : Late childhood.



Loss of function of :

1. Na^+Cl^- cotransporter.
2. Transient receptor potential M-6 (TRPM6) : Responsible for absorption of magnesium.

Consequences :

1. Na^+, Cl^- lost in urine.

↓

Decrease in intravascular volume.

↓

Rening angiotensin aldosterone system activated.

↓

Active space

- Na^+ absorption
- K^+ excretion

↓

Hypokalemic hypochloremic metabolic alkalosis

2. Loss of magnesium ⟶ Hypomagnesemia

Bartter syndrome	Gitelman syndrome
• Hypokalemic hypochloremic metabolic alkalosis. • Blood pressure normal. • Hypercalciuria. • In some cases : Increased level of prostaglandins in Kidney.	• Hypokalemic hypochloremic metabolic alkalosis. • Blood pressure normal. • Hypomagnesemia.

Treatment of Bartter and Gitelman syndrome :

- Correction of electrolyte imbalance.
- Bartter syndrome : Indomethacin (Prostaglandin inhibitor) may be effective.

Liddle syndrome 00:17:25

- Inheritance : Autosomal dominant.
- Mutation : SCNN1B and SCNN1G gene.

- Gain of function mutation : Epithelial sodium channel (ENaC) in collecting duct.
- Consequence : Increased absorption of sodium into blood

↓ Hypertension ↓ Aldosterone suppressed

↓ K^+ loss in urine ↓ Hypokalemic alkalosis

- Also known as pseudo-hyperaldosteronism :
  - Hypertension.
  - Hypokalemic alkalosis.
  - Low aldosterone levels.

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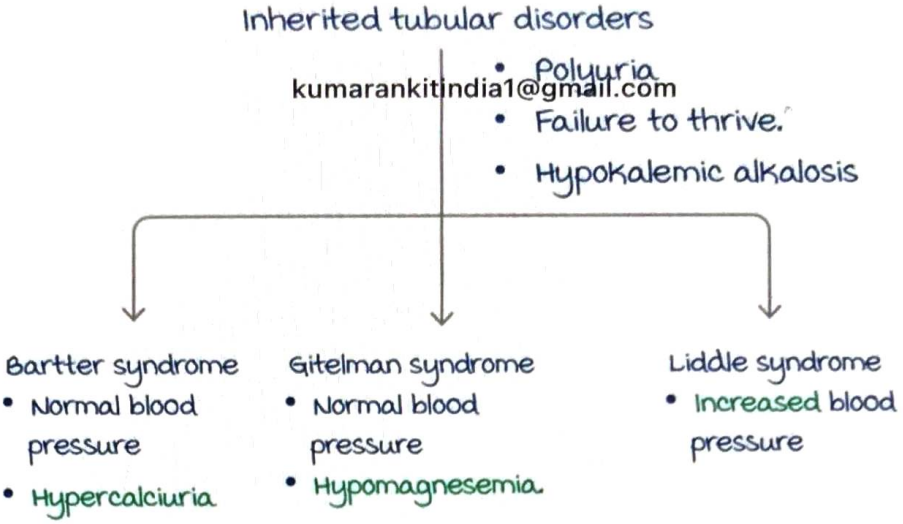
- Treatment : Amiloride (ENaC Blocker)



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RENAL INSUFFICIENCY

Other terms for renal failure :

- Acute renal failure : Acute kidney injury.
- Chronic renal failure : Chronic kidney disease.

Hemolytic uremic syndrome 00:00:58

2nd most common cause of acute renal failure.
1st most common cause : Decreased perfusion to kidney.

Cytotoxin mediated injury :
Initial event is infection : Infection caused by bacteria producing shiga toxin.
Western : Shigella dysentriae Type 1
Southern parts of asia /India : E.coli (ETEC) : O 157; H7.
Recent epidemic : O 104; H4.

Mode of infection : Ingestion of unpasteurized food or undercooked meat.

Etiology :

Toxin producing enteropathogens.

↓

Endothelial damage

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↓

Thrombus in blood vessel.

Traumatic lysis of RBCs (hemolysis) : microangiopathic hemolytic anaemia (MAHA).	Thrombus in Intrarenal blood vessels. ↓ Renal dysfunction → AKI	Lysis of platelets : Low levels of platelets.

most common type of HUS following diarrhoea : D+ HUS.
Rare : D− HUS.

Active space

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HUS features & investigations

00:06:40

Features :

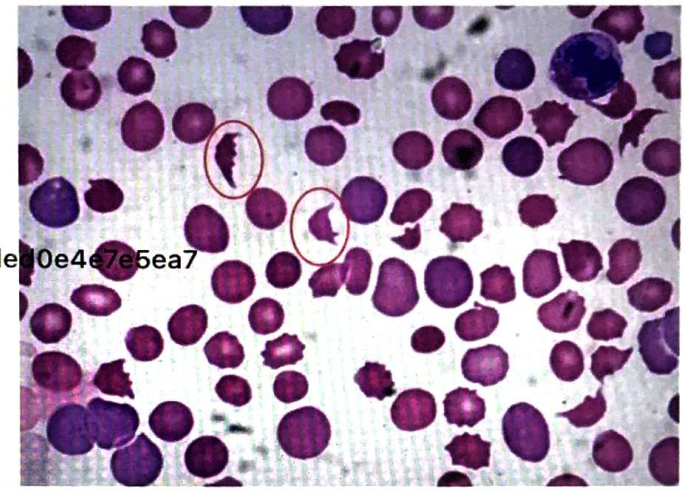
Most commonly occurs in children < 4 years old.

Initial event is diarrhea.

After 5 to 10 days of diarrhoea ⟶ Pallor (due to hemolysis), weakness, oliguria (AKI) develops.

HUS investigations :

Peripheral smear :



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Evidence of hemolysis : Lysed RBCs/schistocytes.

Low platelets.

Other investigations :

- Elevated blood urea & serum creatinine.
- Coomb's negative (not mediated by antibodies).

Definitive testing :

- Stool culture to isolate the bacteria.
- PCR to detect the bacterial antigen.

However, the initial infection would have subsided by the time HUS develops. Hence stool culture would be negative.

Hence a definitive way would be to demonstarate presence of shiga toxin by ELISA.

Active space

HUS treatment 00:09:41

HUS is a self limiting condition.

- Treatment : Supportive.
- Antibiotics : No role in management.
  If antibiotics are given, it may worsen the course as antibiotics can lyse the bacteria present, causing further release of preformed toxins which worsens the disease.
- Dialysis in severe HUS.

Atypical HUS/ D (-) HUS 00:11:28

- Not preceded by diarrhoea.
- Caused by complement defect :

Deficiency of factor H or antibodies to factor H.

↓

Unregulated activation of complement pathway.

↓

Thrombosis.

↓

Hemolysis, low platlets, AKI.

- Triggered by minor illness like respiratory illness.

Management :

- Plasmapheresis : Helps in removing antibodies to factor H.
  Plasma infusion (FFP) : Provide factor H ⟶ Decrease activation of factor H.
- Anti C5a monoclonal antibody : Eculizumab (inhibits further complement activation).

Acute Kidney Injury 00:10:46

Definition by Kidney disease improving global outcomes (KDIGO) guidelines :

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1. S. Creatinine
   - Increase in S. creatinine by >/= 0.3mg/dl from baseline within 48hrs.
   - Increase in S. creatinine to >/= 1.5 times from base line within past 7 days.

2. Urine output
   - Urine output </= 0.5ml/kg/hr for >/= 6hrs.

Staging of Acute kidney injury

00:13:32

	S. Creatinine	Urine output
Stage 1	↑ 1.5 – 1.9 times (baseline)	< 0.5ml/kg/hr for 6–12hrs
Stage 2	↑ 2 – 2.9 times (baseline)	< 0.5ml/kg/hr for >/= 12hrs
Stage 3	↑ >/= 3 times (baseline) or S. Creatinine >/= 4mg/dl Any patient on Dialysis	< 0.3 ml/kg/hr for >/= 24hrs or Anuria >/= 12hrs

Schwartz formula : Calculation of GFR in children.

$$\text{Estimated GFR (eGFR)} = K \times \frac{\text{length (cm)}}{\text{S. creatinine (mg/dl)}}$$

K = constant

- < 1 year : 0.45
- > 1 year/ Adolescent girls : 0.55
- Adolescent boys : 0.7

Active space

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Acute kidney injury causes

00:17:45

Pre Renal	Renal	Post Renal
m/C cause of AKI in children. Hypoperfusion : • Dehydration • Blood loss • Congestive cardiac failure	Parenchymal : • Glomerulonephritis • HUS (2nd m/C) Tubular : • Acute tubular necrosis (ATN) • Acute interstitial nephritis/necrosis (AIN) Vascular : • Renal vein thrombosis.	Obstruction • Stones • Tumors

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Management of AKI

00:20:33

1. Fluid management
   - Pre - renal : Normal saline 20-30ml/kg over 45-60 minutes.
     Response : Diuresis 2ml/kg over 2-3 hrs urine output.
     If No response : add Furosemide 1-2mg/kg.
   - Renal : Fluid restriction determined by :urine output
     Insensible water loss(300 - 400ml/m^2)

     Monitoring by :
     - Daily weight chart :
       Daily decrease of 0.5 - 1% body weight.
     - Serum Sodium : Remain in normal range
       (135 - 145 mEq /L)
2. Anemia :
   - It is usually due to hemodilution in AKI and gets corrected along with fluid correction.
   - Blood transfusion if anemia is due to blood loss.

Active space

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- Blood transfusion if anemia is due to blood loss.

3. Electrolyte imbalance
   - metabolic acidosis
   - Dilutional hyponatremia

- Hyperkalemia : S. Potassium > 5.5 mEq/L.

Serum potassium > 6 mEq/L :

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Potassium binding/ Exchange resins

Kayexalate – Sodium polystyrene sulfate.

Serum potassium > 7mEq/L : Predisposed to arrhythmias.

- Stabilize myocardium : 10% calcium gluconate (0.5 – 1ml/kg) over 5–10 minutes under cardiac monitoring.
- Intracellular shift of potassium :

  Insulin – glucose drip.

  $NaHCO_3$.

  Nebulization (Salbutamol).

  ↓ if all this fails

  Persistent hyperkalemia → Dialysis

Chronic Kidney Disease 00:31:06

Definition of CKD :

- GFR < 60ml/ min/ 1.73m^2 for >/= 3 months or
- Persistence of structural or biochemical anomalies (example : Proteinuria, hematuria) abnormalities in the kidney for >/= 3 months.

National Kidney foundation – Kidney disease outcome quality initiative (NKF-KDOQI) Staging :

Active space

Stage	Description	GFR (mL/min/1.73 m^2
1	↑ GFR (Hyperfiltration) Early stage : Surviving nephrons and blood vessels hypertrophy and proliferate to increase kidney function.	>/= 90
2	mild ↓ GFR Later stages : surviving nephrons affected.	60-89
3	moderate ↓ GFR	30-59
4	Severe ↓ GFR	15 - 29
5	Kidney failure	<15 or on dialysis

m/c cause of CKD in children : Structural anomalies of kidney

Management of CKD

00:36:18

Nutrition : Normal diet.

Anemia :

- It generally occurs when GFR < 40ml/min/m^2
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- Normocytic normochromic anemia
- m/c cause in CKD : ↓ Erythropoietin
- Treatment : Darbepoietin subcutaneous injection
  (Decreases need for blood transfusion.

Bone changes :

CKD is a high bone turnover disease due to increased activity of osteoblasts & osteoclasts.

- 1 α hydroxylation occurs in kidney to yield active form of vitamin D
- Vitamin D deficiency and low calcium → ↑ PTH
  ↓
  Bone resorption.

Active space

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- Osteitis fibrosa cystica –
  Hypocalcemia
  Hyperparathyroidism
  Hyperphosphatemia (as kidney is not able to excrete phosphate)

Treatment :

- Calcitriol supplementation.
- Calcium supplementation.
- Phosphate binders : Calcium acetate, calcium citrate, Sevelamer (non calcium based binder).

Short stature in CKD :

Indication for recombinant growth hormone (rGH) treatment.

Hypertension :

- Dietary sodium restriction : <2g/ day.
- Anti-hypertensive medication of choice :
  1st line : ACE inhibitors.
  2nd line : ARB (angiotensin receptor blockers).

If all the above measures fail, start dialysis.
Best treatment : Renal transplantation.

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Active space

GROWTH HORMONE DEFICIENCY AND HYPOTHYROIDISM

Growth hormone deficiency and hypothyroidism

00:00:16

Growth hormone deficiency :

1. Isolated : Only GH deficient.
2. Hypopituitarism : GH deficient along with other pituitary hormones.

Causes :

Overall MC cause : Idiopathic GH deficiency.

1. Congenital :

Developmental :

Septo-optic dysplasia.

Ectopic pituitary.

Genetic mutations:

GH/GHRH (Growth hormone releasing hormone) genes.

2. Acquired:

- Trauma.
- Tumors : Craniopharyngioma, germinoma.
- Infiltration of pituitary : Histiocytosis, sarcoidosis.
- Irradiation : Radiation induced pituitary necrosis.

m/c etiology : Idiopathic

Clinical features:

Child appears normal at birth (as GH has role only after birth).

Presentation starts in infancy.

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- Growth retardation (short stature) : Proportionate short stature (upper and lower segment ratio normal for age), bone age decreased, delayed dentition.
- Appearance : Doll like facies (fullness of cheeks), over weight/obese (Low GH → No lipolysis → Fat deposition in cheeks and body).

Leave Feedbac

If GH is deficient as a part of Hypopituitrism :

- micropenis : Low gonadotropins
- Hypoglycemia : Low ACTH
- Congenital hyperbilirubiemia/jaundice

Investigations in GH deficiency 00:09:02

Screening test :

IGF I levels (Insulin Like Growth Factor I).

IGF BP 3 (Binding protein) levels :

Less sensitive.

Preferred in children with malnutrition (IGF levels altered by nutritional aspects).

Confirmatory test :

- Serum GH levels :
  Unreliable if checked randomly, as GH is secreted in a pulsatile manner from Pituitary
- GH stimulatory test/GH provocative test :
  Stimulate pituitary → GH released → Estimate.
  Substance used for this test : Insulin, Arginine, Clonidine.

GH < 10 ng/mL : Confirmatory for GH deficiency.

Treatment of GH deficiency :

Recombinant GH (rGH) : Given as S/C injection.

Given till child reaches desirable levels.

Indications of rGH :

Mnemonic : GTCS in Prader Willi syndrome :

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GH deficiency.

Turner's Syndrome.

CKD associated short stature.

SGA (> 2 years); SHOX gene mutations.

Idiopathic short stature.

Noonan syndrome.

Prader Willi syndrome.

Laron's dwarfism 00:15:45

Defect : GH receptor defect (resistance to GH action).

Short stature.

GH levels increased.

Active space

Diagnosis : IGF levels decreased.

Treatment : mecasemin (rIGF - IGF I replacement)

Hypothyroidism

00:18:16

Congenital hypothyroidism :

Very common.

In India, 4 in 1000 patients affected.

most common preventable cause of intellectual disability in children.

Etiology :

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1. Anomaly in thyroid gland (thyroid dysgenesis) :
   - Ectopia of thyroid gland.
     m/c location of ectopic thyroid : Lingual thyroid.
   - Aplasia of thyroid gland.
   - Hypoplasia of thyroid gland.

   Sporadic in nature.

   No associated goiter.

2. Thyroid dyshormonogenesis :

   m/c defective step : Defect in organification (Thyroid peroxidase deficiency).

   Inherited condition : Autosomal Recessive.

   Diffuse goiter seen. Soft in consistency.

3. Iodine deficiency (Endemic goiter) :

   Uncommon in India now.

   Underdeveloped countries : Still seen.

4. Maternal autoantibodies (TSH receptor blocking antibodies) :

   Transplacental transfer.

   Transient condition.

5. Pendred syndrome :

   Autosomal recessive.

   Defect in chromosome 7 → Pendrin gene → Cl^- transporter.

   Sensorineural hearing loss (Pendrin gene also in cochlea).

Active space

Leave Feedba

Features of congenital hypothyroidism 00:26:40

1st week after birth → most babies asymptomatic (mother's thyroxin protection).

After 1st week :

- Decreased activity/ lethargic.
- Hypothermia/cold skin.
- Delayed passage in meconium (constipation).
- Coarse facies.
- Hoarse cry.
- Macroglossia.
- Umbilical hernia.
- Prolonged jaundice (> 2 weeks after birth).
- No goiter is seen.

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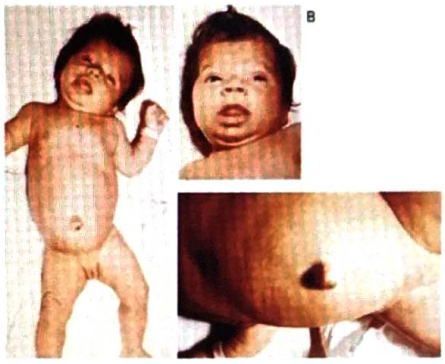


Investigations in hypothyroidism :

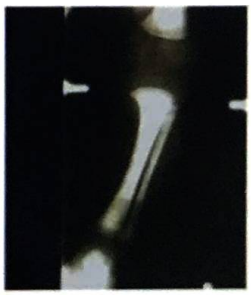
TFT : Decreased T3, T4; Increased TSH.

Newborn screening of hypothyroidism : Increased levels of TSH.

- TSH invariably high in all babies when done immediately after birth.
- Therefore, done between days 2-5 (day 3).

X-ray :

- Delayed osseous maturation → Delayed bone age.
- Epiphyseal dysgenesis/ stippled epiphysis (fragmented epiphysis).
- Skull : Intra-sutural bones/wormian bones.



Diagnostic approach to congenital hypothyroidism

Active space

Radionucleotide study (thyroid scan):

Iodine 123 > technetium 99. Thyroid scan:

No uptake → USG thyroid —No gland→ Thyroid agenesis

↓ Gland normal

Check for TSH receptor antibodies

+ve: Maternal antibodies.

-ve: Iodine trapping/TSH receptor defect.

Ectopic uptake → Ectopic thyroid.

Increased uptake → Dyshormonogenesis/Iodine deficiency.

Treatment of hypothyroidism 00:37:37

Oral thyroxine supplementation (T4):

Start at the earliest (prevent any brain abnormalities).

Starting dose: 10–15 micro grams/kg/day.

Response: Increase in T4 by 1 week, normalization of TSH by 1 month.

Adjust the dose.

Lifelong necessity.

Endemic cretinism:

- Severe iodine deficiency.
- Common in underdeveloped countries.
- It is of two types

1) Neurological type:
   - most severe.
   - Very low IQ.
   - Goiter seen.
   - Spasticity.
   - Deaf mutism (sensorineural hearing loss).

2) Myxedematous type:
   - Facial puffiness.
   - Periorbital puffiness
   - Low IQ
   - Loss of lateral 1/3rd of the eyebrows.
   - Goiter not noted.

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Active space

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Acquired hypothyroidism :

Autoimmune condition : Hashimoto's thyroiditis (anti-TPO antibodies).

Age group : Adolescent females.

Features :

- Decrease in growth velocity (short) - first sign
- Constipation.
- Dry skin.
- Weight gain despite poor appetite.
- Goiter: Firm and nodular.
- Delayed puberty.

Treatment:

Oral T4 supplementation: 10-15 micrograms/kg/day.

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Active space

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Leave Feedbac

ADRENAL DISORDERS IN CHILDREN

Cushing's syndrome

00:00:17

↑ Glucocorticoids in the body.

Etiology : Exogenous causes >> endogenous causes.

Exogenous causes : Due to prolonged intake of steroids.

Endogenous causes : Due to increased production from the adrenal cortex and can be of two types as follows :

1. ACTH dependent (due to increased ACTH) :
   a) Pituitary adenoma : Cushings disease.
   b) Ectopic source from a tumor :
      - Neuroblastoma (most common).
      - Wilm's tumor.
      - Carcinoid tumor.
2. ACTH independent (due to adrenal pathology).
   For example : Carcinoma or adenoma of the adrenal gland.

Cushing's syndrome (CS) :

Clinical Features :

Common feature in children : Growth retardation (characterised by decreased growth velocity and short stature).

- Moon facies : Due to fullness of the cheeks.
- Buffalo hump : Due to deposition of excess fat in the back of neck.
- Abdominal/truncal obesity : Due to abnormal fat distribution.
- Purplish abdominal striae : Due to distension of the abdomen.
- Hypertension : Because glucocorticoids have some intrinsic mineralocorticoid activity.
- Hirsutism in females : It is seen only in ACTH dependant CS because of increased production of testosterone from adrenal gland.
- Delayed puberty.

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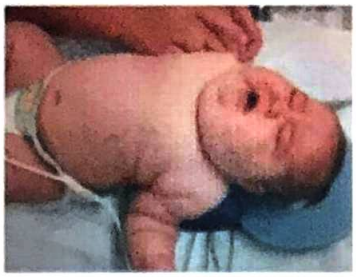
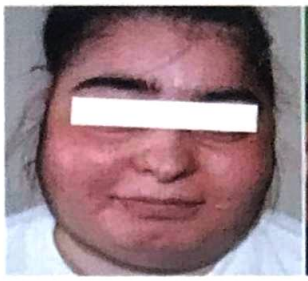
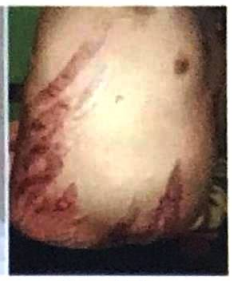
- Muscle weaknes : Proximal group of muscles are affected more.



Moon facies





Purplish abdominal striae

Cushing's syndrome management

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00:08:38

Screening test (to find the evidence of increased steroid in the body).

- 24 hour urine free cortisol measurement.
- Overnight Dexamethasone suppression test :
  Overnight administration of single dose of 0.3mg/m^2 Dexamethasone followed by measurement of cortisol levels in the morning.
  The levels of cortisol remain high in CS.

Confirmatory test :
Low dose Dexamethasone suppression test : 5 microgram/kg of Dexamethasone is given every 6 hours for 2 days and then the levels of cortisol are estimated.
The levels of cortisol remain high in CS.

Serum ACTH (to determine if it's an ACTH dependent or an independent CS) :

1. Low ACTH (< 5 pg/ml) : Adrenal cause/ACTH independent.
2. High ACTH (> 15 pg/ml) : ACTH dependent CS.
   a) Pituitary adenoma.
   b) Ectopic ACTH secreting tumor (very high level of ACTH > 100 pg/ml).

Another test to differentiate pituitary vs ectopic cause of CS:
High dose Dexamethasone suppression test :

Active space

If there is suppression of ACTH : Pitutary cause.
If there is no suppression of ACTH : Ectopic cause.

Cushing's syndrome : Treatment 00:14:02

Depends on the cause of CS.
If due to any tumor : Surgical removal of the tumor.
Medical management (medical adrenalectomy) :
Drugs used are :

- Ketoconazole.
- metyrapone.
- mitotane.

Adrenal insufficiency 00:15:17

Etiology : Primary causes are more common than secondary cause.

Primary adrenal insufficiency (problem in the adrenal gland) :

1. Most common cause : Autoimmune destruction of the adrenal gland, Addison's disease (AD).
   AD can occur as an isolated entity or as a part of Autoimmune Polyendocrine Syndrome (APS).
   AD is usually associated with :
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   a) APS 1 : AD + chronic candidiasis + hypoparathyroidism.
   b) APS 2 : AD + autoimmune thyroid disease (Hashimoto's thyroiditis) and/or type 1 DM.
2. Infections :
   a) HIV.
   b) Tuberculosis.
   c) Acute meningococcemia : There is sudden adrenal haemorrhage.
   This is called Waterhouse Friderichsen syndrome.
3. Metabolic causes :
   a) Adrenoleukodystrophy.
   b) Congenital Adrenal Hyperplasia (eg.; 21-hydroxylase deficiency).

Active space

Leave Feedba

Secondary adrenal insufficiency (problem in the CNS) : Tumor or trauma associated with pituitary gland causing decreased production of ACTH.

Features and management of AI (it depends on whether it is acute or chronic) :

- Acute presentation is most commonly due to acute meningiococcemia.
- Chronic presentation is mostly due to AD.

Acute presentation features :

- Shock (fluid unresponsive shock).
- ↓ Aldosterone : Hyponatremia, hyperkalemia.

Treatment : IV fluids + Inj. Hydrocortisone.

Chronic presentation :

- ↓ Glucocorticoids :
  1. Hypoglycemia.
  2. Lethargy.
- ↓ mineralocorticoids :
  1. Postural hypotension.
  2. Salt craving.

ACTH stimulation test : ACTH stimulation → 60 mins → low cortisol levels (< 18 microgram/dL).

Treatment : Hydrocortisone (glucocorticoid) + Fludrocortisone (mineralocorticoid).

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If the cause is due to autoimmune condition then, the drugs is given lifelong.

Steroid requirement increases at the time of stress, so the dose of steroids given should also be increased :

- For a minor stress (infections) : Dose should be increased 2-3 times.
- For a major stress (surgery) : Dose should be increased 4-5 times.

Active space

CAH/ Congenital Adrenal Hyperplasia 00:26:15

It is an autosomal recessive disorder.

Characterised by the deficiency of the enzymes in the adrenal biosynthesis of hormones.

Common enzyme deficiencies :

- 21- Hydroxylase (21-OH) deficiency (MC).
- 11-Hydroxylase (11-OH) deficiency.
- 17-Hydroxylase (17-OH) deficiency.
- 3 β-Hydroxy Steroid Dehydrogenase (3 β HSD) deficiency.

Pathophysiology :

↓ synthesis of adrenal hormones (especially glucocorticoids).

↓

Feedback stimulation of the pituitary adrenal axis.

↓

Increased production of ACTH (has a trophic effect/growth on the adrenal glands).

↓

Stimulation of the adrenal gland.

↓

Hyperplasia of the adrenal gland.

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Active space

Leave Feedback

Steps of adrenal gland biosynthesis of steroid hormones :

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Active space

21-hydroxylase deficiency 00:29:46

↓ Aldosterone :

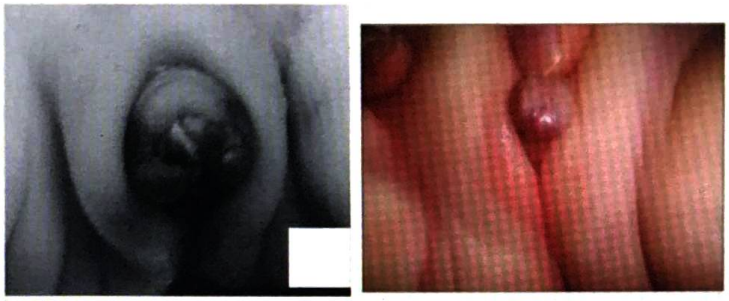
- Low BP (hypotensive shock).
- Hyponatremia (salt wasting type of CAH).
- Hyperkalemia.

↓ Cortisol : Hypoglycaemia.

↑ Testosterone:

- In female baby : Ambiguous genitalia.
- In male baby : Precocious puberty.

↑ ACTH : Hyperpigmentation (as ACTH is similar to MSH/ Melanocyte Stimulating Hormone).



Ambiguous genitalia in a female child

Treatment:

- Lifelong Hydrocortisone + Fludrocortisone (doses of these need to be increased during stress).
- For female babies : Corrective surgery like clitoroplasty is done within 2-6 months of age.

Newborn screening test for early detection of CAH : Estimating the levels of 17-OH progesterone (17-OHP), which will be elevated in this CAH.

11β-Hydroxylase deficiency 60c6b3eeaa8ded0e4e7e5ea7 00:36:19

↑ Testosterone : Ambiguous genitalia in female and precocious puberty in male babies.
11-deoxycorticosterone (has mineralocorticoid activity) levels are elevated, hence, blood pressure is actually increased in this condition although aldosterone is deficient.

↓ Cortisol : Hypoglycemia.

↑ ACTH : Hyperpigmentation of skin.

Active space

Leave Feedbac

17α-Hydroxylase deficiency 00:38:45

↑ Aldosterone : High BP, hypernatremia, hypokalemia.

↓ Cortisol : Hypoglycemia.

↓ Testosterone :

In male baby – Undervirilisation :

1. Hypospadias.
2. Bifid scrotum.
3. Cryptorchidism.
4. Severe deficiencies : Ambiguous genitalia.

In female baby : Normal genitalia.

At the time of puberty, delay in the development of secondary sexual characteristics.

↑ ACTH : Hyperpigmentation.

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3β-HSD deficiency 00:42:09

↓ Aldosterone (Low BP).

↓ Cortisol (Hypoglycemia).

↓ Testosterone.

↑ ACTH (Hyperpigmentation).

↑ DHEA (Dehydroepiandrosterone) : It is a weak androgen.

In female baby : Features of mild virilisation like hirsutism, acne, menstrual irregularity.

In male baby : Features of undervirilisation like hypospadias, Bifid scrotum, Cryptorchidism.

CAH : Simplified approach 00:44:34

Salt wasting features like low BP, hyponatremia, shock.

- If present : 21-OH deficiency, 3β–HSD deficiency.
- If absent : 11-OH deficiency, 17-OH deficiency.

Based on testosterone levels :

- If increased in cases like Precocious puberty in male and ambiguous genitalia in a female : 21-OH deficiency, 11-OH deficiency.
- If decreased then 17-OH deficiency, 3β HSD deficiency which can present with normal genitalia in a female baby or under virilized in male or rarely ambiguous genitalia.

Active space

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PUBERTAL DISORDERS

Onset of puberty :

- Girls : 8 to 12 years
- Boys : 9 to 13 years

Types of puberty and cut offs : based on appearance of secondary sexual characteristics.

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	Female	Male
Precocious puberty (Advanced)	< 8 years	< 9.5 years
Delayed puberty	> 13 years	> 14 years

Delayed puberty

00:01:22

Overall most common cause : Constitutional delay (Central).

HCG / GnRH stimulation test

↓

Brisk response : Increased Testosterone / Estrogen ⟶

Only in case of constitutional delay.

Causes :

Central	Peripheral
↓	↓
Hypogonadotropic Hypogonadism (Decreased LH, FSH)	Hypergonadotropic Hypogonadism
Syndromes :	Syndromes :
1. Kallman syndrome (KAL 1) Anosmia	1. Turner syndrome
2. Prader Willi syndrome	2. Klinefelter syndrome
3. Laurence Moon Biedl Bardet syndrome.	3. Premature ovarian failure
4. Trauma/ tumor in pituitary.	4. Males only : (Testosterone)
5. Infection.	• 5 α reductase deficiency.
6. Infiltrative disorders : Histiocytosis, sarcoidosis.	• Androgen insensitivity syndrome (AIS).

Active space

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Treatment : Supplementation of hormones.

Hormone replacement : Wait till 12 years (girls)
14 years (boys)
To avoid early fusion of epiphysis and reduction in height.

Precocious puberty

00:12:24

Advanced puberty :

Causes

Central (Gonadotropin dependent)

- More common.
- Idiopathic in girls.
- Associated with tumors in boys.
  MC : Hypothalamic hamartoma
  Gelastic seizure :
  Inappropriately laughing.
- Damage to pituitary
- Malformations :
  1. Arachnoid cyst.
  2. Hydrocephalus.

Peripheral (Gonadotropin independent)

- Gonads :
  Functioning tumours (ovary and testis) :
  Seminoma, Germinoma.
  McCune Albright syndrome.
- Adrenal :
  Tumors
  Boys : Congenital adrenal hyperplasia
  Enzyme deficiencies :
  21- OH 11- OH

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McCune Albright syndrome :

Autonomic endocrine hyperfunction of 1 or more glands like thyroid, pituitary, adrenal, ovary.

- Gene defect : gain of function mutation of GNAS1 gene which codes for α subunit of Gs protein which results in uncontrolled activation of the receptor leading to increased production of hormone from endocrine glands .
- Triad : Precocious puberty (increased estrogen from autonomic activity of ovary also termed as peripheral precocious puberty),

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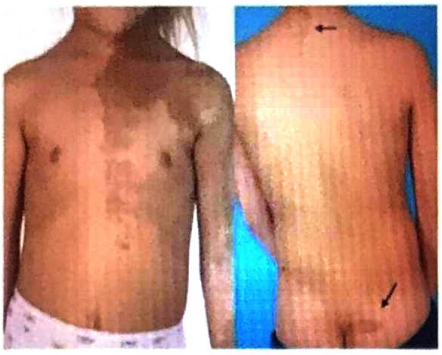
↓

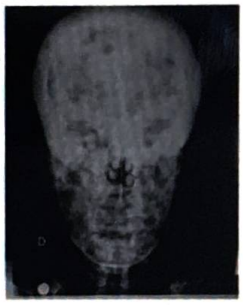
can enlarge and result in torsion ← ovarian cysts



Café au lait macules : Characteristic hyperpigmented macule, single.

Mc Cune Albright syndrome : Irregular borders, Coast of Maine appearance.

Neurofibromatosis : Regular borders, Coast of California appearance.



Polyostotic fibrous dysplasia : Lytic expansile lesions that can present in multiple bones.

MC : Base of skull & proximal part of femur.

Can cause bowing of the femur : Shepherd's crook deformity.

Other manifestations : Hyperthyroidism
Excess growth hormone.

Levels of FSH and LH decreases.

Treatment :

Aromatase ionhibitors (Letrozole) Inhibits the last step of estrogen synthesis thereby preventing the progression of puberty in children.

Investigations :

- Biochemical : LH (better indicator), FSH assays.
- Radiological imaging : Bone age (advanced)
- Imaging : If central Precocious puberty we have to rule out a possible cause like hypothalamic hamartoma.

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If its a peripheral precoccious puberty then we have to look for tumors in testis or ovary.

Active space

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Treatment :

- GnRH analogues (Central precocious puberty) :
  Leuprolide
  ↓
  Desensitization and down regulation of GnRH receptors.
  ↓
  Decrease in the release of LH and FSH.

Normal variants

00:23:39

Premature thelarche (breast budding) :

- Occurs : 1 – 2 years.
- No other signs of secondary sexual characters or puberty will develop after.
- LH, FSH : Normal.
- Bone age : Normal.

Premature pubarche (appearance of pubic hair & axillary hair) :

- Also known as Premature adrenarche.
- Familial occurence.

Premature menarche :

- Local cause : Infection.
  Foreign body.
  Sexual abuse.

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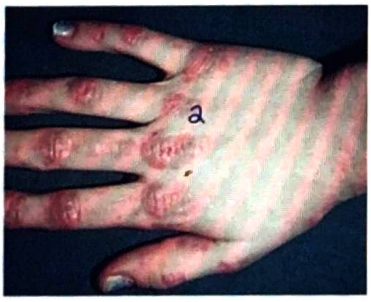
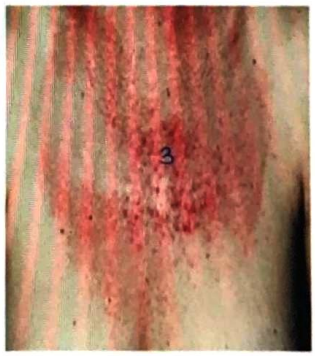
RHEUMATIC DISEASES OF CHILDHOOD

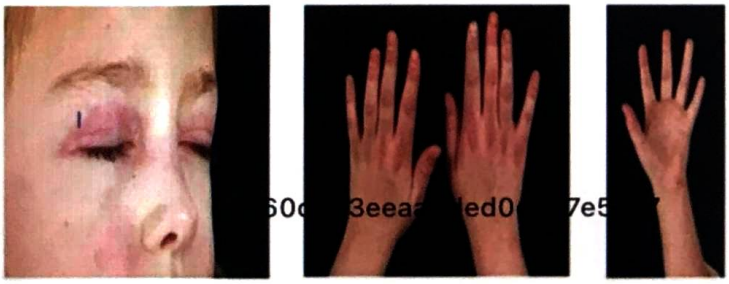
Juvenile dermatomyositis 00:00:20

It is most common Inflammatory myositis in childhood.
Muscle + skin manifestations.
Skin manifestations :

- Heliotrope rash (blue violet erythematous rash around eyelids) + periorbital edema.
- Gottron's papules (erythematous, scaly red lesions over dorsal surface of joints : MCP, interphalangeal joints).
- Shawl sign (associated feature) : Erythematous rash over neck & upper anterior chest region.
- Mechanic hands : Thick erythematous scaly lesions over the fingers and toes.







Heliotrope rash (1), Gottron's papules (2), Shawl sign (3)

Active space

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Diagnostic criteria :

Skin features(Heliotrope rash and Gottron papules) + any 3 of the following :

1. Symmetrical + proximal muscle weakness (shoulder/thigh). Gower's sign positive (using hands for supporting thigh to get up).
2. Elevation of muscle enzymes (muscle fibers inflamed and destructed) → Creatine Kinase, Aldolase.
3. EMG → Short motor unit potentials and fibrillations.
4. muscle biopsy → Necrosis /inflammation of muscle fibers.

Serology findings :

1. Myositis associated antibodies (MAA) (non specific) : SSA, SSB, Anti-dsDNA.
2. MSA (Specific) : Anti Jo I, Anti MIA.

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Treatment :

1. Corticosteroids (IV > oral).
2. methotrexate (maintenance treatment/ long term) for 2 years.

Juvenile idiopathic arthritis (JIA) 00:10:22

Arthritis : Intra articular swelling , and any two of the following :

- Limitation in the joint movement.
- Tenderness/pain while moving joint.
- Warmth of joint (any 2 at least).

It is most common rheumatological arthritis in children.

Onset : < 16 years.

Duration : ≥ 6 weeks.

coomon cause of increased childhood morbidity like school abseentism,emotional disturbances.

Subtypes of JIA :

- Oligoarticular JIA: ≤ 4 joint (m/c subtype).
- Polyarticular JIA : ≥ 4 joints. Both Rheumatoid factor(RF) positive and negative.
- Systemic JIA (s JIA). (F=m).
- Psoriatic JIA.
- Enthesitis related JIA. (m > F).

Active space

All other JIA : F > M.

Oligoarticular JIA : Age group is 3 to 5 years.
≤ 4 joint affected in the first 6 months of disease.

Subdivided into :

- Persistent : Always </= 4 joints at any time of disease.
- Extended : > 4 joints affected after 6 months of disease.
  m/c affected joints : Knee > Ankle > Wrist.
  Involvement of hip joint, small joints of hands and feet : Rare.

Extra-articular manifestations : Chronic uveitis which may lead to blindness (especially if ANA positive).
HLA-DR 5/8 positivity is a pre disposition for Oligo articular JIA.

Polyarticular JIA :

	RF negative	RF positive
	HLA DR-B1.	HLA DR-4, DR-1.
Clinical picture	Knee , hip, wrist	Large joints and small joints of hand and feet, cervical spine, temporomandibular joint (TMJ).
age	Any age.	Late childhood and early adolescence (9 to 11 years).
Symmetry	Asymmetrical involvement.	Symmetrical involvement.
Association	ANA positive → Chronic uveitis.	Rheumatoid nodules (noticed in extensor surfaces of joints like elbow and occiput, Achilles tendon)

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Systemic JIA :

Clinical features :

1. Fever.
2. Any one of the following:
   - Rash.
   - Hepatosplenomegaly.
   - Lymphadenopathy.
   - Serositis (pleuritis/pericarditis).

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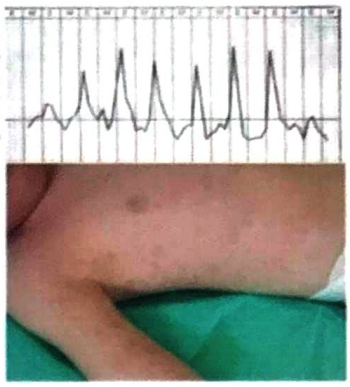


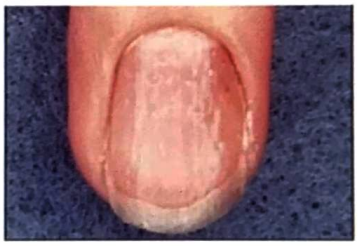
Evenings → Intermittent fever spikes (1 to 2 per day/ Quotidian fever) → Rash in trunk only during fever (non pruritic, maculopapular, salmon pink colored transient rash).

Psoriatic JIA :

Arthritis before /together and after Psoriasis.

Clinical features :

1. Psoriasis.
2. If psoriasis not evident, any one of the two :
   - Nail pitting.
   - Dactylitis.
   - First degree relative with psoriasis.



Pitting in Psoriatic arthiritis

Enthesitis related arthritis :

- Inflammation of tendons at the site of attachment to the bone.
- Males are more commonly affected females.
- Onset : Older children > 8 years.
- Joint : Lower limb joints (hip/knee/ankle).
- Asymmetrical involvement.
- HLA B-27 positive.
- Associated with sacroilitis.

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Investigations of JIA :

It is a clinical diagnosis usually.

Active space

1. CBP : Anemia of chronic disease + increased WBC + increased platelets.
2. X-rays : Normal in early stages.
   Later stages :
   - Soft tissue swelling around joint.
   - Peri-articular osteopenia.

   most sensitive investigation for joint involvement : MRI.
3. Serology (antibodies) for prognostic importance.

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Treatment of JIA :

1. NSAIDs (Pain control) : Naproxen, Ibuprofen.
2. Intra-articular steroids (Triamcinolone) if not controlled with NSAIDs.
3. Disease modifying agents (methotrexate, Hydroxychloroquine).
4. Biological agents :
   - Interleukin 1/IL-1 receptor antagonists : Anakinra.
   - Anti TNF alpha : Infliximab, Etanercept, Adalimumab.
   - IL-6 receptor antibodies : Tocilizumab.
   - Inhibitors of T-cell activation : Abatacept.

Henoch Schonlein purpura 00:35:00

It is Small vessel vasculitis of childhood.
IgA deposits in vessels → IgA vasculitis.
Also called as Purpura rheumatica and leukocytoplastic vasculitis (debris of neutrophils around blood vessels).
Affects the age group : 4 to 8 years.

Features :

1. Purpura (non-thrombocytic, palpable, bleeding spots over lower limbs or buttocks) doesn't affect upper part of the body .
2. Arthralgia/arthritis.
3. Post prandial abdominal pain.
4. Renal manifestations (nephritis) → Developed later, initially normal.

Skin Biopsy → IgA deposition.
Diagnosis : Purpura + any one of the 4 features given above.

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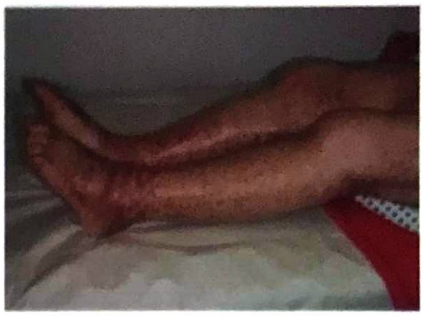
Investigations :

- Platelet count : Normal.
- Initial urine analysis : Normal.
- Follow up needed for 6 months to
- look for later renal manifestations.
- Prognosis : Excellent.



Complications :

- Acute : GI (Intussusception).
- Chronic : Chronic Kidney disease.

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Kawasaki disease

00:42:00

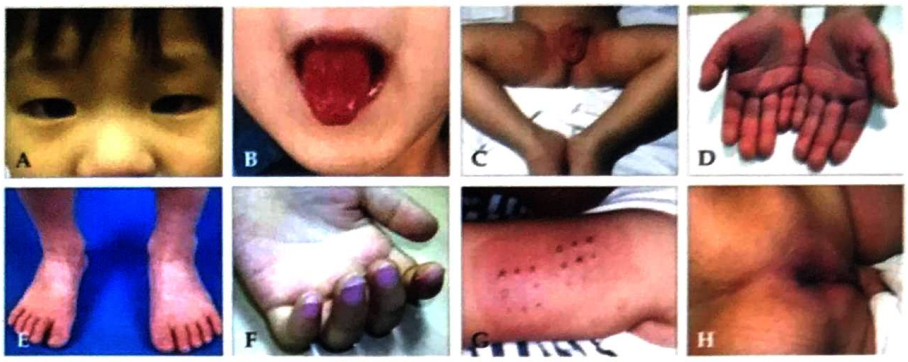
- Medium vessel vasculitis.
- M/c vasculitis of childhood in India.
- Age group : < 5 years.
- Etiology : Idiopathic/unknown.

Three phases :

	Acute phase	Subacute phase	Convalescent /recovery phase
Duration	1 to 2 weeks	3 to 4 weeks	6 to 8 wks
Features	Prolonged fever > 5days. Other manifestations: Mnemonic CREAM : Conjunctivitis : Bilateral, non purulent. Rash : Nonspecific maculopapular rash. Extremities : Erythema and edema, Periungual desquamation (subacute). Adenopathy (lymphadenopathy : usually Single, cervical). Mucosal involvement : Cheilitis/glossitis (strawberry tongue).	Periungual desquamation (peeling of the nails). Thrombocytosis.	Recovery from illness

Active space

Compli-cations	Acute: myocarditis (50-70% incidence)	Coronary artery aneurysms (20-30%) Increased risk of mortality if size ≥ 8 cm.	



A : Conjunctivitis, B : Strawberry tongue, E : Arthiritis, F : Periungual desquamation, G : Rash due to reactivation of BCG scar.

Diagnostic criteria of Kawasaki disease :

Fever + At least 4/5 features of CREAM.

If < 4 criteria → Incomplete Kawasaki disease → Echocardiography → If coronary arteries involved, treat like Kawasaki disease.

Other features :

- Reactivation of BCG scar. (G)
- Peri anal desquamation. (C, H)
- USG abdomen → Hydrops/distended gall bladder.
- Sterile pyuria.
- Aseptic meningitis.
- Arthritis.
- Hepatitis.

Complications :

- Acute : myocarditis (50-70 %)
- Subacute : Coronary aneurysm (20-30 %). Increased risk of mortality due to thrombosis and rupture (size ≥ 8mm).

Treatment of Kawasaki disease :

1. DOC : IVIG (Intravenous immunoglobulin : 2 gm/kg infusion)
2. High dose aspirin (80 to 100 mg/kg).

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- Anti-inflammatory.
- Continued till patient afebrile for 48 hours, then we can decrease dosage.

3. Low dose Aspirin (3 to 5 mg/kg/day).
   - Continued till 6 to 8 weeks.

After 6 to 8 weeks, Echocardiography has to be done :

- If ECHO Normal : Stop Aspirin.
- Coronary abnormalities : Continue Aspirin till normalized.

Active space

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APPROACH TO ANEMIA IN CHILDREN AND NUTRITIONAL ANEMIA

Anemia in children

00:01:00

WHO definition of anaemia :

Based on hemoglobin (Hb) levels as per the age group.

Age	Cut off hemoglobin (g/dL)
6 months to 5 years	11
5 years to 11 years	11.5
≥ 12 years	12

Approach to anaemia :

History :

- Diet (nutritional deficiency).
- History of chronic disease.
- History of hemolytic disorder : Jaundice.

•Family history of :

Anaemia / Jaundice.

Gall bladder disease.

Splenectomy.

Physical examination :

Findings	Associated conditions
Associated congenital anomalies	Fanconi's anaemia
Petechiae / Purpura	Leukemia, Aplastic anaemia, Hemolytic Uremic Syndrome/ HUS
Generalized lymphadenopathy	Leukemia, systemic onset Juvenile Idiopathic Arthritis (JIA)
Splenomegaly	Leukemia, hemolytic disorders.

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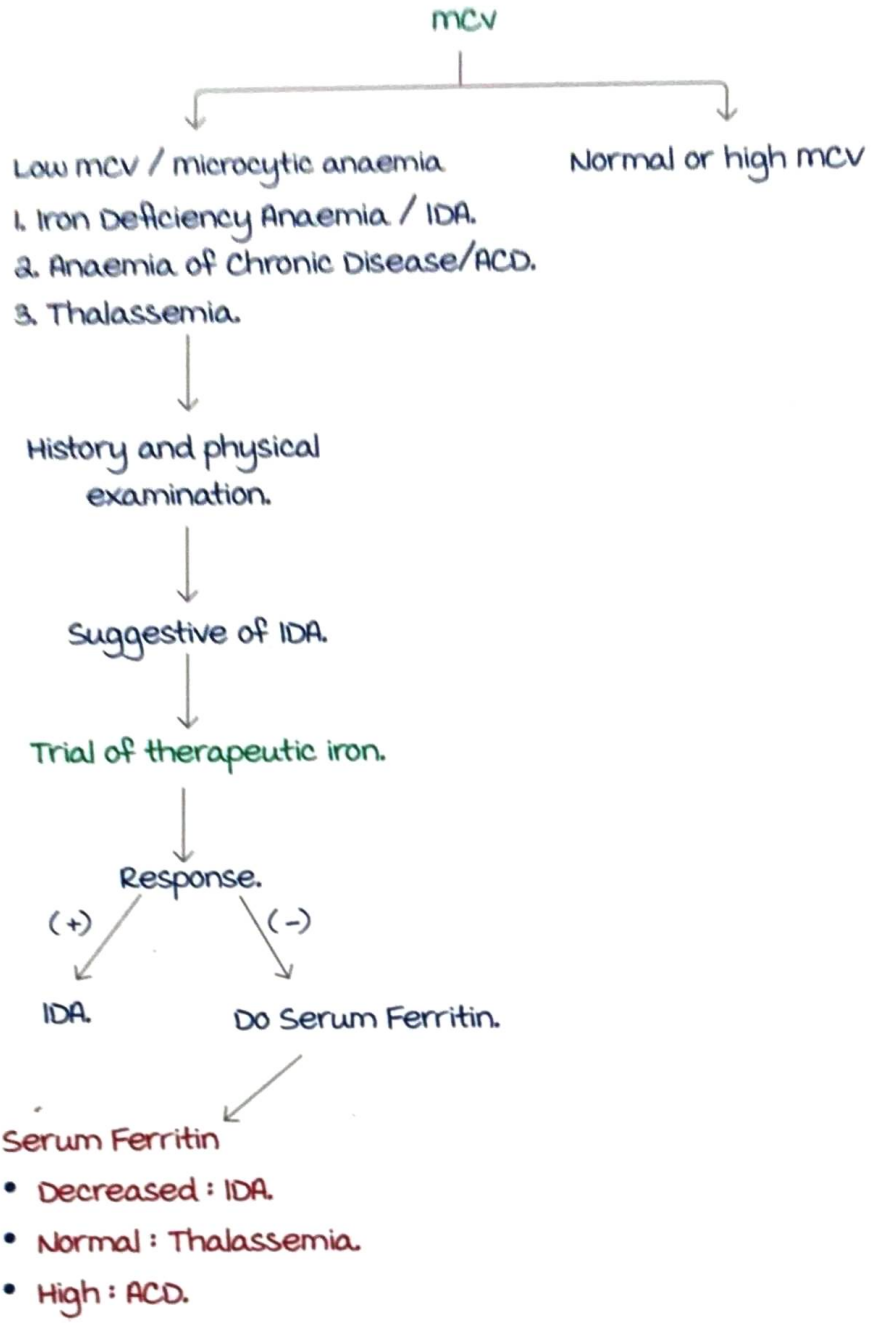
Lab approach to anaemia :

Mean Corpuscular Volume/ MCV.

Normal range : 90 ± 8 fL.



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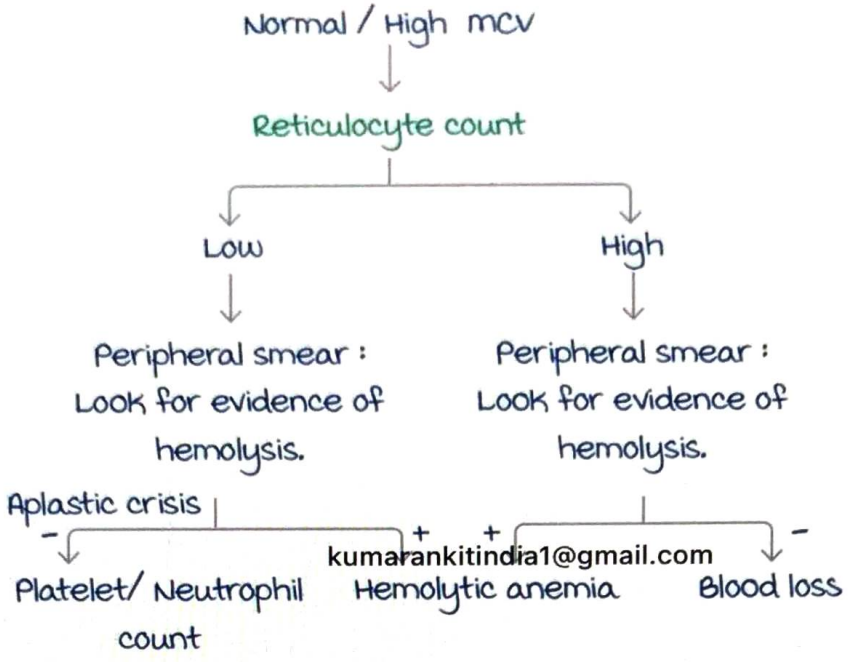

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Low :
1. Bone marrow failure
2. Megaloblastic anaemia

Normal :
Pure Red cell aplasia
Eg : Blackfan Diamond syndrome

The aplastic crisis is facilitated by infections especially Parvovirus B19 infection.

Iron deficiency anemia

00:16:00

Most common condition causing anaemia in children worldwide : Iron deficiency.

Iron deficiency : State of insufficient iron, such that it's unable to maintain normal physiological function. It is characterized by low levels of iron stores / ferritin.

Risk factors of iron deficiency in children :

- Dietary deficiency of Iron.
- Increased demand : Growth spurts (first 2 years & puberty).
- Preterm / low birth weight (LBW), VLBW < 1.5kg : Low iron stores.
- Malabsorption : Celiac disease (Villous atrophy in Small intestine).
- Hookworm infestation (blood loss and decreased absorption treated by Deworming).

Age group : 9 months to 24 months.

Active space

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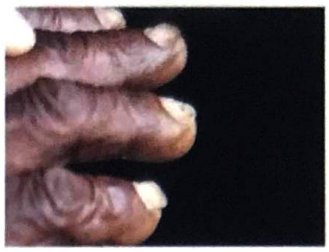
In the first 6–9 months : No anemia as iron stores are derived from the mother during the last trimester of pregnancy.

Clinical features :

- Pallor, irritability, fatigue.
- Koilonychia : Spooning of nails.
- Platynychia : Flat nail.
- Glossitis, angular stomatitis.



Koilonychia and Platynychia

Associations in clinical situations :

- PICA.
- Temper tantrum.
- Breath holding spells.
- Restless leg syndrome.

Lab features :

Microcytosis, anisocytosis.

Low MCV.

Low serum ferritin.

Treatment :

Most economical and effective form of iron is

ferrous sulfate : 20% elemental iron.

Dose : 3 to 6 mg/kg/day.

Duration : 4 to 6 months (replenish iron stores).

Response following iron therapy :

Time after iron administration	Response
12 to 24 hours	Subjective improvement. Replacement of intracellular iron enzymes.
36 to 48 hours	Bone marrow response : Erythroid hyperplasia (Increase in erthroid cells).
48 to 72 hours	Reticulocytosis : Peaks at 5 to 7 days.
4 to 30 days	Increase in hemoglobin.
1 to 3 months	Replenish iron stores.

Active space

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Megaloblastic anaemia

00:27:55

Macrocytes : Delay in nuclear maturation.

Causes : Deficiency of Vitamin B12 and Folic acid.

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Vitamin B 12 deficiency causes :

1. Diet : Pure vegetarian / Vegan.
2. Pernicious anaemia : Autoimmine antibody mediated damage causing deficiency of Intrinsic factor needed for absorption of Vitamin B 12
3. Malabsorption :
   - Crohn's disease.
   - Chronic pancreatitis.
   - Diphyllobothrium latum (fish tapeworm).
4. Metabolic :
   - Homocystinuria.
   - Orotic aciduria.
   - Methyl Malonic Aciduria / MMA.

Folic acid deficiency causes :

- Diet : Excess goat milk.
- Malabsorption : Giardia.
- Decreased utilization of drugs :
  - 6 mercaptopurine.
  - Methotrexate.
  - Trimethoprim.

Clinical features :

- Pallor.
- Smooth beefy red tongue.
- Hepatosplenomegaly (30- 40%).
- Hyperpigmentation (Knuckles).
- Neurological (Vit B12 deficiency), can precede anaemia :
  - Loss of position / Vibration sense : Earliest.
  - Unsteady gait, paresthesia.

Lab features of peripheral smear :

- Macro ovalocytes (large size RBCs).
- Hypersegmented neutrophils.

Treatment :

Folic acid :

1 to 5 mg daily for 3 to 4 weeks.

Active space

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Vitamin B12 :
1000 mcg oral.
Daily for 2 weeks.
Weekly for the next 2 weeks.
Monthly for a lifetime.

Inherited pancytopenia 00:37:36

Decrease in RBCs, WBCs and platelets due to bone marrow failure.

Fanconi's anaemia :
Inheritance : Autosomal recessive.
Defect in DNA repair (Chromosomal breaks seen in cells).
Features :
Pallor, Increased risk of infections, petechiae/ purpura.
3 "S" :

1. Short stature.
2. Skeletal anomalies :
   Like radial ray defect : Absent or hypoplastic radius/ thumb.
3. Skin pigmentation :
   1. Hyperpigmentation.
   2. Cafe-au-lait macules.
4. Microcephaly + tapering jaw.

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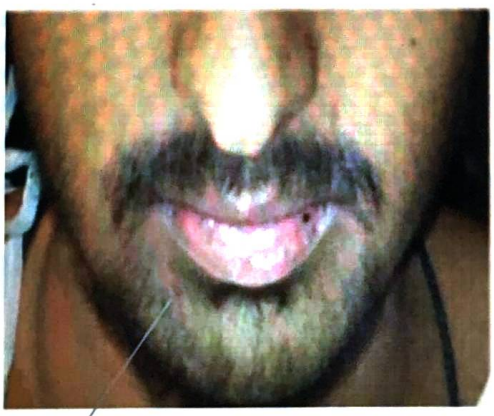
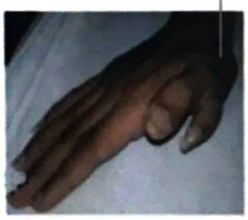


Tapering jaw



Radial ray defects
bifid thumb

Chromosomal breaks

Treatment : HSCT (Hematopoietic Stem Cell Transplantation).
Palliative : Oral androgens.

Long term follow up due to the increased risk of AML, and oral/ liver cancers.

Active space

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CONGENITAL HEMOLYTIC ANEMIA

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Congenital hemolytic anaemias 00:00:10

3 categories :

- Defect of the red cell membrane. E.g. : Hereditary spherocytosis.
- Hemoglobinopathies. E.g. : Alpha / beta thalassemia, sickle cell anaemia.
- Disorders of red cell metabolism. E.g. : Glucose 6 phosphate dehydrogenase deficiency.

Hereditary spherocytosis 00:01:22

Mode of inheritance : Autosomal dominant in 75% cases.
Others : Autosomal recessive.
De novo mutation.
Basic defect : Cytoskeleton defect (defect in RBC shapes).
Defective proteins :
Ankyrin : Most common.
Spectrin : Most severe.
Band 3 : Adults.

Pathophysiology :

Spherical RBCs.
↓ Decreased deformability.
Not able to squeeze through splenic microcirculation.
↓
Trapped / Engulfed by splenic macrophages.

Features :
Clinical : Anemia, intermittent jaundice, splenomegaly.
Neonatal jaundice in 50%.
Family history positive for jaundice / splenectomy.

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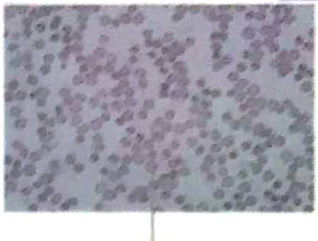
Lab :
Peripheral smear shows :
Densely stained spherocytes.



Spherocytes

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Increased MCHC / mean corpuscular haemoglobin concentration.

Increased osmotic fragility (incubated osmotic fragility) :
Noted especially when incubated for 24 hours at 37°C.

Management :
Folic acid supplementation for a life.
To decrease the possibility of red cell hypoplasia.

Splenectomy :
Normalise RBC life span / Improve anaemia.
Is deferred until 5 years, as it increases post splenectomy infections as encapsulated organisms are destroyed in spleen :

1. Pneumococci.
2. H. influenza type b.
3. Meningococci.

All children need to be vaccinated for these infections at least 2 weeks before undergoing splenectomy.
Penicillin prophylaxis is given for 1 year post splenectomy.

Hemoglobinopathies 00:11:08

1. Sickle cell anaemia.
2. Thalassemia.

Sickle cell anemia 00:11:30

Mode of Inheritance : Autosomal recessive.

Active space

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Defect :

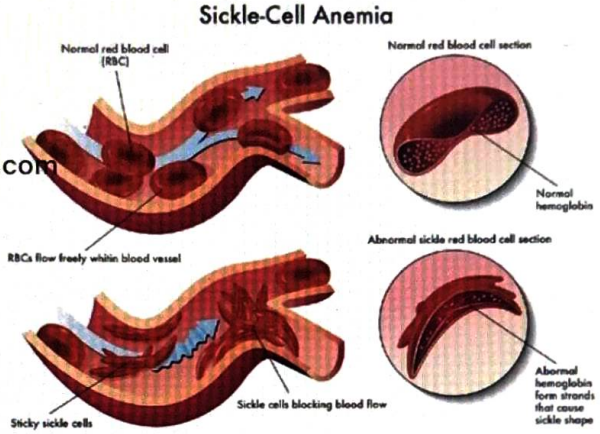
Glutamic acid in 6th position of β globin chain.
↓ Replaced by
Valine

Mnemonic : Glutamate Goes, Valine walks.

Reason : Point / missense mutations.
Pathophysiology :

Type of Hemoglobin : HbS (Sickle shaped).

Hypoxia.
Dehydration.
Acidosis.
↓
Sickling of RBCs.
↓
Blockage of blood vessels.



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Clinical features :
Icterus, pallor, and mild splenomegaly.
increased risk of recurrent Febrile episodes.

Vaso occlusive crisis :
1. Bony pain : Commonly affects the long bones (Femur, tibia).
The first manifestation of sickle cell anemia in Infants :
Dactylitis.
Dactylitis : Painful swelling of hands and feet.

2. Kidney : Renal papillary necrosis.
Hence the inability of kidneys to concentrate urine results in isosthenuria : The osmolarity of urine is the same as plasma / Fixed specific gravity.

3. Spleen : Repeated infarctions due to multiple vaso occlusive crises.

Active space

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Results in auto splenectomy at 6 years (Asplenia).

4. CNS : Stroke.

Acute chest syndrome :
(Features resembles sudden pneumonia)
Symptoms :
1. Fever.
2. Cough.
3. Tachypnoea.
4. Hypoxemia.

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Chest X-ray : New infiltrates.

Treatment :
1. O2 support.
2. I.V fluids.
3. Bronchodilators.
4. Antibiotics : 3rd generation cephalosporins + Macrolides.

Sequestration crisis :
Sickled RBCs block the venous outflow of the spleen.
↓
Engorgement of spleen.
A sudden increase in the size of the spleen.
Decline in Hb > 2g. (Increased destruction of RBCs).

Aplastic crisis :
Sudden decline in all cell counts : Pancytopenia.
Precipitated by Parvovirus B19.

Lab features :
1. Sickled RBCs.
2. Sickling test : 2% sodium metabisulfite.
3. Hb electrophoresis : HbS.
4. Gold Standard : HPLC / High-performance Liquid Chromatography (for all hemoglobinopathies).

Management :
Hydration and analgesia (Narcotics).

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Red cell transfusions : To improve O2 carrying capacity in aplastic / sequestration crisis.

Exchange transfusion : Stroke and acute chest syndrome.
Goal : HbS < 30%.
Drugs :
Hydroxyurea (in > 9 months age) to HbF :
Decreases pain, crises, hospitalisation, and transfusion requirement.
L-Glutamine : Decrease vaso occlusive crisis.
HSCT /Hematopoietic stem cell transplant is curative.

Thalassemia

00:28:55

Reduction in the synthesis of one or more haemoglobin polypeptide chains. Types α, β Thalassemia.

β Thalassemia :
β globin chains are coded by 2 genes on Chromosome 11.
The mutation is point mutation.
Most common : Splicing mutations.

β Thalassemia Spectrum :
1. β Thalassemia trait : Asymptomatic, mild anaemia,
HbA2 – 8% and HbF – 5%.

2. β Thalassemia Intermedia : Anaemia, jaundice, splenomegaly.
- Transfusion ± (infrequent if required).

3. β Thalassemia major : Transfusion dependent, most severe.
Colley / mediterranean anaemia.
(Thalassa : Sea, emia : related to blood).

Pathophysiology :
Absence of β globin chains.
A relative excess of alpha chains (insoluble).
↓
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Accumulate in erythroid precursors (Bone marrow).
↓
Erythroblast undergo apoptosis.

Active space

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↓

Ineffective erythropoiesis

↓

Anaemia

↓

Regular blood transfusions are required.
This predisposes to iron overload.

Iron overload in the heart ⟶ CCF / Congestive cardiac failure.
Cause of Death : CCF.

Extramedullary hematopoiesis :
Compensation for ineffective erythropoiesis which causes the following :

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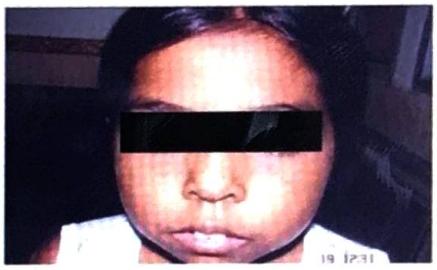
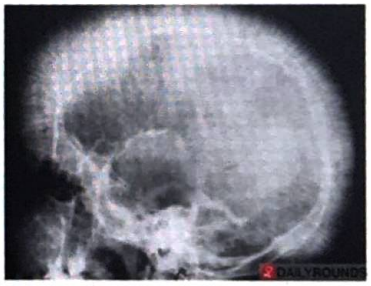
Chipmunk facies :
Frontal bossing.
Prominence of cheeks (maxilla).
Dental malocclusion.



Chipmunk facies

X-Ray skull : Expansion of skull.
Hair on end appearance / Crew cut appearance.

Hepatosplenomegaly.



Crew cut appearance

Management :
Require blood transfusion is required :
Interval 3 to 4 weeks.
The goal is to maintain Pre- transfusion Hb level.
(9.5 to 10.5g/dL) for optimal growth ⟶ decrease deformities.

The best assessment of iron overload.

↓

MRI Liver – R2/R2*
MRI Heart – T2W.

Managed by Iron chelation :
Iron overload occurs after 1 year of transfusion therapy or Ferritin > 1000ng/ml.

Active space

Iron chelators :

1. Deferoxamine (S.C / I.V).
2. Deferasirox (Oral) : more effective in decreasing organ damage.
3. Deferiprone (Oral).

Oral drugs are more commonly used now a days.

Curative treatment :

Hematopoietic Stem Cell Transplant / HSCT : 90% cure rate.

Needs to be done before liver involvement (Hepatic fibrosis).

Red cell metabolism :

RBCs lack mitochondria.

Anaerobic metabolism of glucose for ATP.

Pathway : HMP shunt.

Rate limiting enzyme : G6PD / Glucose 6 Phosphate Dehydrogenase.

Role of G6PD :

Provides : NADPH and Glutathione.

Glutathione protects RBCs from oxidative stress.

Disorders of red cell metabolism :

G6PD deficiency.
↓
Low levels of Glutathione.
↓
RBCs undergo oxidative stress.
↓
Hemolysis.

G6PD deficiency

00:47:54

Inheritance : X linked recessive.

more frequent in persons of African, Mediterranean, and Asian ancestry.

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Clinical presentation :

Intermittent episodic hemolysis.

Factors precipitating hemolysis :

1. Infections.
2. Food : Fava beans.

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3. Drugs :
   - Primaquine.
   - Sulfonamide (Sulfamethoxazole).
   - Phenazopyridine (Pyridium).
   - Nitrofurantoin.
   - Nalidixic acid.

Lab features :

Heinz bodies : Denatured haemoglobin seen as intracellular inclusion bodies in RBCs.

G6PD deficiency
↓
Oxidative stress on RBCs
↓
Hb ⟶ methaemoglobin.
↓
Denaturation.
↓
Intracellular inclusions : Heinz bodies.

Bite cells :

These abnormal RBCs
↓ Pass through spleen.
Splenic macrophages bite chunks of RBC containing inclusion bodies.

Management :

Most episodes of hemolysis are self limiting.
Avoid precipitating factors.
Early diagnosis and treatment is recommended.

Active space

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BLEEDING DISORDERS

Bleeding disorders – introduction

- If there is a cut in the skin leading to bleeding, the changes which happen to stop bleeding are :

Vasoconstriction (Vessels) → Platelet plug (Formation) → Coagulation (Clotting factors)

- Defects :
  - Hereditary hemorrhagic Telangiecstasia (Osler – weber – Rendu)
  - Thrombocytopenia
  - Platelet function disorders
  - Hemophilia
  - Scurvy
  - Vasculitis
- Bleeding disorders approach

Finding	Disorders of coagulation	Disorders of platelets or vessels.
– Petechiae	–	+
– Deep dissecting hematmas	+	–
– Superfi cial ecchymoses	Single; Large	multiple; Small
– Hemarthrosis	+	
– Delayed bleeding	+	–
– Bleeding from superficial cuts and scratches.		+

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Idiopathic thrombocytopenic purpura 00:04:13

- Also called as immune thrombocytopenic purpura (ITP)
- Antibody mediated
- Usually in age group 1–7 years
- Types : Acute (m.c type)
  Chronic (persists > 6 months)

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Acute ITP

- Typical presentation :

Non - specific
Viral infection
1-4 WEEK → Thrombocytopenia

- Petechiae, purpura in a previously well child
- No splenomegaly, no lymphadenopathy
- Treatment :
  - > 20,000 / μL. No active bleed → Observation (recovers spontaneously)
  - < 20,000 / μL Active bleed → I.V.I.G (Intravenous immunoglobulins) (Prevent destruction of antibody coated platelets)
  - Steroids
  - Anti-D (Rh^+ children)

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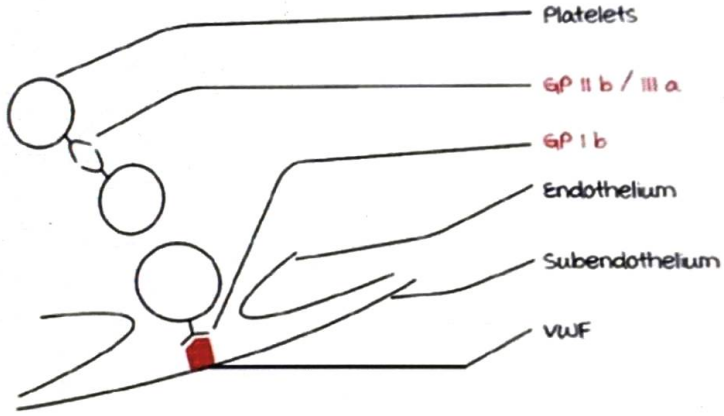
Platelet Function Disorders



- Platelet receptor function :
  - GP IIb / III a → Aggregation
  - GP Ib → Adhesion
- 2 major platelet function disorders :

Glanzmann's thrombasthenia	Bernard soulier syndrome
- Defect in GP II b / III a	- Defect in GP I b
- Defect in platelet aggregation	- Defect in platelet adhesion
- Normal platelet number	- ↑ platelet number
- Normal sized platelets	- sized platelets

Active space

Leave Feedback

Von Willebrand disease

00:12:53

- MC inherited bleeding disorder world wide
- Functions of von willebrand factor (VWF) :
  1. Carrier for factor 8
  2. Helps in attachment of platelet to endothelium
- Types :
  1. Type I
     - ↓ VWF (MC)
     - Treatment : Desmopressin
  2. Type 2
     - Impaired function of VWF
     - Treatment : VWF concentrates
  3. Type 3
     - Absent VWF
     - Treatment : VWF concentrates

Hemophilia

00:16:13

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- Types : Hemophilia A : Factor VIII deficiency
  Hemophilia B : Factor IX deficiency
- Hallmark feature : Recurrent hemarthrosis(bleeding into joints).
- Target joint : Joint prone to recurrent hemarthrosis. Classically, knee or ankle joints are involved.
  Once a joint is affected, it becomes a target joint.
- Treatment : Factor replacement
  Hemophilia A : Amount of factor required : % desired rise x bodyweight (kg) x 0.5
  Hemophilia B : Amount of factor required : % desired rise x body weight (kg) x 1.4
  - % rise is estimated based on severity of bleeding :
    - Severe bleed (CNS) : 100 %
    - Less severe bleed : 30 – 50 %
  - If factor replacement is not available, alternative treatment options :
    - Hemophilia A : FFP/Cryoprecipitate (Does not contain factor IX)
    - Hemophilia B : Only FFP.

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HEMATOLOGICAL MALIGNANCIES

Acute Lymphoblastic Leukemia (ALL) 00:00:13

- ALL : most common type of leukemia in children and most common
- malignancy overall in children.
- Peak age : 2 to 5 yrs
- Boys > Girls
- Risk factors :
  - A – Ataxia telangiectasia
  - B – Bloom's syndrome
  - C – severe Combined Immunodeficiency
  - D – Down syndrome
  - E – Environmental factors (like radiation exposure
  - F – Li Fra[illegible] anemia)
- Types of ALL :
  - (i) progenitor B cell (pre B- cell) : 85% cases
  - (ii) T- cell
  - (iii) mature B- cell
- Features of ALL :
  - (i) Bone pain
  - (ii) Decrease in cell count : Anemia, thrombocytopenia, leukopenia
  - (iii) Infiltration : Hepatosplenomegaly
    - Lymphadenopathy
    - Mediastinal mass → SVC Syndrome (more commonly associated with T- cell type)
- Diagnosis of ALL :

  Bone marrow aspirate : >25% lymphoblast

Active space

Standard risk and high risk

00:05:28

	Standard risk	High risk
• Age	2- 10 yrs	< 1 yr, > 10 yrs
• Gender	Female	male
• Features :		
• Organomegaly	Absent	Present
• mediastinal mass		
• CNS features		
• Initial WBC count	<50,000/cu.mm	> 50,000/cu. mm
• Subtypes	Pre - B cell	mature B -cell
• Genetic factors	Hyperdiploidy, Trisomy 4, 10 .	Hypodiploidy, t (9 ; 22) - Philadelphia t (4 ; 11) - < 1 yr - Infantile leukemia

Intermediate risk - T - cell type of ALL

Treatment of ALL

(i) Induction phase : Prednisolone
Asparginase
Anthracycline
Vincristine

In high risk : Minimal Residual disease - present at the end of induction phase

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- Minimal Residual Disease - Minimal number of blasts present even at the end of induction phase.
  - Done using flow cytometry, PCR.

(ii) Prophylatic CNS therapy :
Intrathecal methotrexate

(iii) Consolidation phase (Intensification phase) :
Methotrexate
Epipodophyllotoxin
Cyclophosphamide
Cytosine - Arabinoside

Active space

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(iv) Maintenance phase
- Methotrexate
- 6 - mercaptopurine

Acute myeloid leukemia (AML)

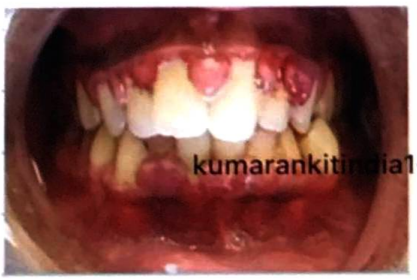
00:13:36

- 2nd most common type of leukemia in children.
- Diagnosis of AML :
  Bone marrow aspirate : >20 % myeloblasts.
- Types of AML :

FAB	Name	Genetic variation and prognosis
M_0	Dedifferentiated / minimally differentiated	Deletion of 7q - poor prognosis
M_1		
M_2	Myeloblastic without maturation	t (8 ; 21)
M_3		t (15 ; 17)
M_4	Myeloblastic with maturation	Inversion - 16
M_5		
M_6	Promyelocytic	
M_7	Myelomonocytic Monocytic Erythroleukemic Megakaryocytic	Down's syndrome - Good prognosis

Good prognosis

- Features of AML :
  Features similar to ALL
  Except : no lymphadenopathy , no hepatosplenomegaly .
  But, M_4 AND M_5 have associated hepatosplenomegaly .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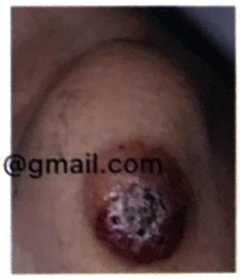
Gum hypertrophy



Chloroma

M_3 : increased risk of DIC
M_4 : Gum hypertrophy
Accumulation of myeloperoxidase : Chloroma

Active space

Leukemia and Down's syndrome 00:18:38

- Down's syndrome has increased risk of leukemia.
- Most common leukemia overall – ALL
- Most common leukemia in age < 3 yrs – AML
- Variant in newborn period :

  Transient myeloproliferative disorder :

  Blasts ⊕ in circulation.

  Hepatosplenomegaly

  GATA – 1 mutation

  20% cases develop leukemia.

Hodgkin lymphoma 00:20:28

Age group :

Bimodal age distribution, 15 –19 years (adolescent age) and > 50 years (5th or 6th decade).

Male predominance in children. This is not observed in older population.

Risk factors :

Infections : HHV 6, CMV, EBV (EBV confers 4 fold high risk of developing HL).

Autoimmune disorders : SLE, ITP (Immune/Idiopathic Thrombocytopenic Purpura).

Clinical presentations :

1. Lymphadenopathy : Cervical group is usually affected.

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Painless.

Firm and rubbery in consistency.

Discrete/matted lymph nodes.

2. Constitutional symptoms : Grading B or B symptoms.

Fever of >38 °C.

Drenching night sweats.

Weight loss >10 % in the preceding 6 months (significant weight loss).

Presence of constitutional symptoms : Grading B

Absence of constitutional symptoms : Grading A (good prognosis).

Active space

Leave Feedback

Presence of constitutional symptoms confers a poor prognosis and indicates the need for aggressive treatment.

3. Asymptomatic mediastinal mass usually involving the anterior mediastinum.

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Types of HL

00:25:58

Based on the pathological features of HD.

Classical type	Nodular/ lymphoyte predominant type
CD 15+ & CD 30+.	CD 25+ & CD 45+
1. Nodular sclerosis : Overall MC type. Females > males. Not associated with EBV. Excellent prognosis. Lacunar variant of RS cell seen.	Not associated with EBV. Excellent prognosis.
2. Mixed cellularity : MC in India. Males > females. Associated with EBV.	modified RS cells : Popcorn cells (fluffy/irregular/blurred appearance), usually seen in nodular lymphocyte predominant type of HL.
3. Lymphocyte rich type : Associated with EBV. Reticular variant of RS cell : Lymphocyte rich type.	
4. Lymphocyte poor/ lymphocyte depleted type : Associated with EBV (90%) and HIV. Poor prognosis.	

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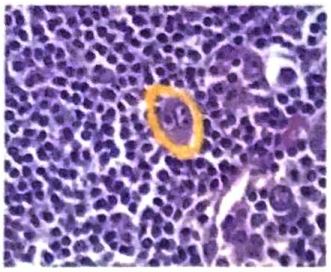
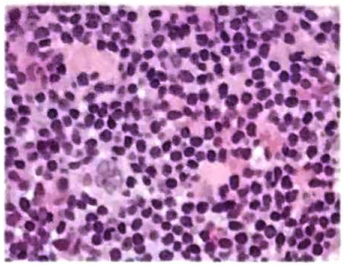
Investigation of choice : Excisional biopsy from the lymph node.



Owl eye inclusion body



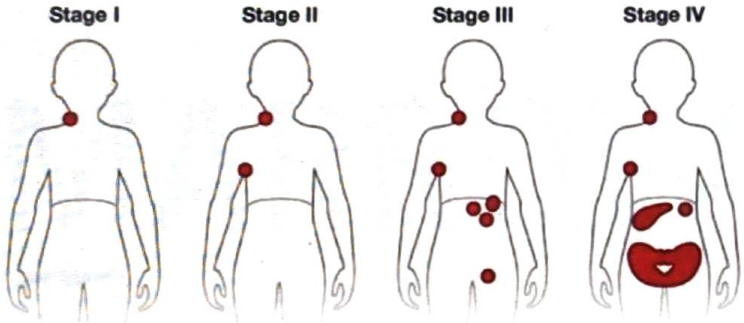
Popcorn type of RS cell

Presence of RS cell/Reed Sternberg cell is the hallmark feature of HL.

Reed Sternberg cells :

It is a germinal center B cell which has undergone malignant transformation with owl eye inclusion body like appearance.

Staging of HL : Modified Ann Arbor staging 00:34:05



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A: Absence of B symptoms B: fever, night sweats, weight loss

©MARROW

Stage 1 : Single LN affected (usually cervical LN).

Stage 2 : ≥ 2 LN affected, only one side of the diaphragm is affected.

Stage 3 : Multiple LN on both sides of the diaphragm.

Stage 4 : Disseminated disease (liver, spleen etc., affected).

A : Absence of B symptoms.

B : Presence of B symptoms.

X : Bulky tumor of > 10 cm in size.

E : Extension into an adjacent extra lymphatic site.

Active space

Leave Feedba

Based on the staging, HL is divided into :

Low risk disease	High risk disease
Stage 1 or 2.	Stage 3 or 4
Absence of B symptoms.	B symptoms are present.
Absence of bulky tumor (X).	Presence of bulky tumor.
Treatment : ABVD regime Adriamycin. Bleomycin. Vinblastine. Dacarbazine.	Treatment : ABVD regimen + Bleomycin + Etoposide + Radiotherapy.

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Non Hodgkin Lymphoma/ NHL

00:18:25

more common than HL.

NHL (60% cases), HL (40% cases).

Age group : 5-15 years.

Malignant proliferation of the lymphocytes T-cells/B-cells/ intermediate cell origin.

Types :

According to the predominance of cells seen in NHL :

B cell type :

- Burkitt's lymphoma (mc type of NHL).
- DLBCL (Diffuse Large B cell Lymphoma).

T-cell type :

- Lymphoblastic lymphoma (overall mc type in India) : It has clinical features like ALL. Associated with increased incidence of intrathoracic/mediastinal tumors.
- Anaplastic large cell lymphoma.

Clinical spectrum of NHL

00:22:25

Characteristic differences between NHL in children and NHL in adults :

Active space

Adults	Children
Localized disease.	Diffusely spread lesions.
Nodal type : Usually involves only LN.	Extranodal : Abdomen, mediastinum, bone marrow.
Low grade.	High grade (rapidly proliferating) lesions.

Salient features of individual types of NHL :

Lymphoblastic lymphoma : Presents as mediastinal/ intrathoracic mass.

Burkitt's lymphoma :

Associated with EBV & malaria, with the following varieties :

1. Sporadic type : MC type.
   EBV association is ≤ 20%.
   Sites : Intra-abdominal tumors.
   Unexplained abdominal pain or distension.
   Right lower quadrant mass.
   >5 years age children : Intussusception (tumor acts as a lead point).
   MC sites : Abdomen > Bone marrow > CNS.

2. Endemic type :
   EBV association is ≥ 95%.
   Endemic in African countries.
   Sites : Intra abdominal tumor & jaw tumors (lytic lesions).

DLBCL (Diffuse Large B cell Lymphoma) : Presents with intra-abdominal mass.

Anaplastic large B cell lymphoma :

Heterogenous condition which presents with unusual sites of infections like :

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Lesions of skin.
Lesions in brain leading to focal neurological deficits.
Unexplained pleural or pericardial effusion.

Active space

Leave Feedbac

St. Jude staging of NHL 00:31:02

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Localized disease (low risk stage) :

Stage 1 : One site (area/node) without involvement of GIT/ mediastinum.

Stage 2 : Multiple LN involvement (on one side of the diaphragm).
Single GIT involvement which is completely resectable.

Advanced type (high risk stage) :

Stage 3 : Multiple sites in the GIT are involved,
non-resectable tumors.
Mediastinal involvement.
Epidural or paraspinal tumor.

Stage 4 : Stage 3 + involvement of bone marrow and/or CNS.

Treatment of NHL :
Cyclophosphamide.
Vincristine.
Prednisolone.
Doxorubicin.

For high risk stages : methotrexate is added.

Langerhans cell histiocytosis/LCH 00:35:21

Also called Histiocytosis X : Mysterious nature of this condition.
Non malignant disorder.
Clonal proliferation of langerhans like cell.
Langerhan cells are the tissue resident macrophages in the epidermis of skin.

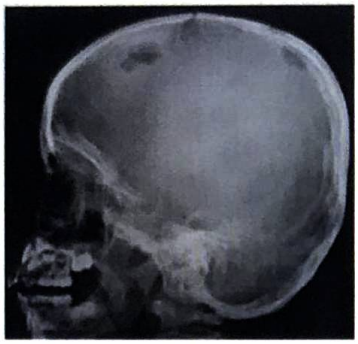
Recent theory :
Proliferation of immature cells of myeloid lineage.

Clinical findings :

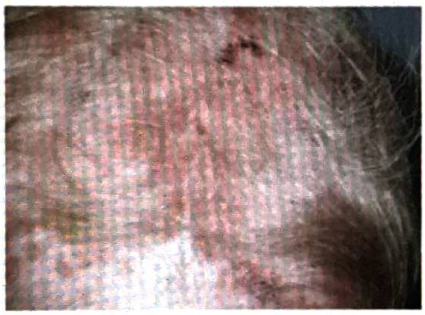
Wide spectrum of disease, depending on the age :

Active space

MC overall site : Bone (lytic lesions), MC bone affected is the skull.
2nd MC site is jaw/mandible : missing tooth/loose tooth.



Skin involvement : Irregular scales over the skin of the scalp.
(seborrheic dermatitis/cradle cap) with discharge. Difficult to treat & differs from classical seborrhoeic dermatitis.



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Infiltration of the pituitary stalk leading to deficiency of vasopressin (diabetes insipidus) and MRI brain shows thick pituitary stalk & loss of bright spot in the posterior pituitary (due to loss of vasopressin granules).

LCH clinical spectrum :

Older children/adolescents : Eosinophilic granuloma (lytic lesions of the skull).

Hand-Schuller-Christian disease : 2-6 years age children, triad of :

- Calvarial defects.
- Diabetes insipidus.
- Exophthalmos.

Letterer-Siwe disease : < 2year children.
Systemic form : Fever, irritability, weight loss, cirrhosis , ataxia, dysarthria.

Active space

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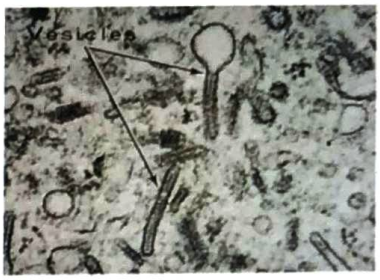
Hashimoto-Pritzker disease : In the neonatal period.
Also called congenital self healing form of histiocytosis.
It is a cutaneous form of LCH.

Diagnosis :
Biopsy specimen.
Immunohistochemistry : CD 1a, CD 207 (langerin positive), S-100 +.
Electron microscopy : Birbeck granules (tennis racquet appearance).

Treatment :
Single lesion :
- Curettage.
- Intralesional steroid therapy.

Multiple lesion :
- Chemotherapy :
  - Vinblastine (mc).
  - Steroids.
  - Mercaptopurine.
  - Etoposide.

Refractory disease : Cladribin.



Birbeck granules

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SOLID TUMOURS IN CHILDREN

Neuroblastoma 00:00:16

It is the most common intra-abdominal tumor in children.
It is also the most common extra-cranial solid tumor in children.
Overall, most common solid tumor in children is intracranial/brain tumors; 2nd most common solid tumors in children is neuroblastoma.
Neuroblastoma usually occurs in infancy, → Considered most common malignancy in infancy.

Cell of origin : Neuroblasts (derived from neural crest cells).

Site of involvement :
1. Adrenal gland (most common) → Often called as suprarenal tumor.
2. Para-vertebral sympathetic chain or ganglia(2nd most common).
3. Posterior mediastinum.
4. Pelvis.

Pathology :
Solid tumor with small, round, uniform blue pattern of cells.
Blue cells are called so due to their scanty cytoplasm along with presence of hyperchromatic nuclei.

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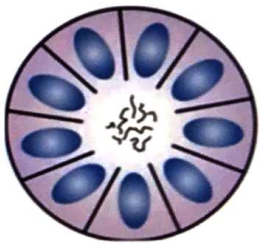
However, this is not diagnostic for neuroblastoma as there are other examples of small round blue cell tumors :

- Non-Hodgkin's lymphoma.
- Rhabdomyosarcoma.
- Ewing's tumor.



Homer-Wright rosettes :
It is made up of individual tumor cells that have rose petal/rosette appearance with eosinophilic material called neuropil in the center.

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Neuropil/neurites are the cytoplasmic processes.

Clinical features :

It is an abdominal tumor and can be presented as an abdominal mass that is firm, fixed with an irregular outline. Usually a midline mass.

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Constitutional symptoms include :

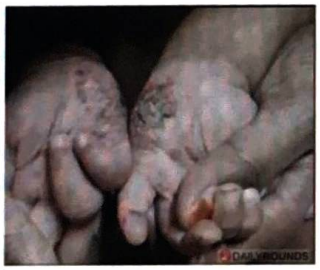
- Fever.
- Weight loss.
- Irritability.

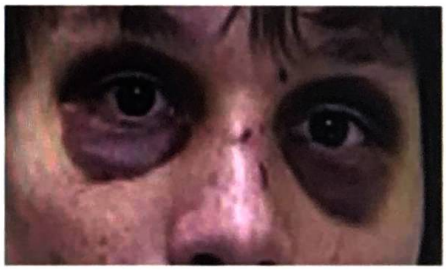
They may have other features like Horner's syndrome or heterochromia iridis :

Horners syndrome	Heterochromia iridis
Due to involvement of sympathetic chain. The child will present with classical symptoms like anhydrosis, miosis & ptosis.	The patient presents clinically with different color of both iris.

Some cases may present with metastasis :

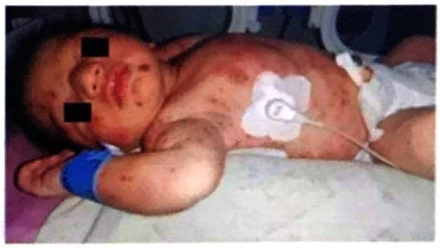
- most common site of metastasis is bone, typically presents with bony pain.
- metastasis to the Retro-orbital region with infiltration often presents as peri-orbital swelling and ecchymosis called 'Racoon eyes'.





Racoon eyes

metastasis in the skin can present as purplish/bluish subcutaneous nodule called 'Blueberry muffin lesions'.



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In some cases neuroblastoma may present with characteristic paraneoplastic manifestations like :

- Opsoclonus-myoclonus ataxia/dancing eye/dancing feet syndrome : Usually associated with the presence of anti-neuronal antibodies.
- VIP (vasoactive intestinal polypeptide) hypersecretion : Intractable watery diarrhea, hypokalemia.

Investigations :

1. Initial Investigations : Urinary catecholamines/urine markers :

   As the tumor is neural crest derivative they secrete catecholamines,

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   metabolites of which are secreted in the urine. Commonly found are :

   VMA (vanillylmandelic acid).
   HVA (homovanillic acid).

   However, they are not specific to neuroblastoma, they may be elevated in tumors like pheochromocytoma.
   Specific urine marker for neuroblastoma : Estimation of levels of NSE (Neuron specific enolase).

2. Diagnostic investigations :

   Imaging studies : CT Abdomen is most used.

   - Presence of Stippled calcifications in CT are characteristic of the tumor. Tumor mass displaces the kidney inferolaterally in the CT, this is useful to differentiate from a tumor arising from the kidney like Wilm's tumor.
   - Radionuclide scanning to rule out metastasis, most used radionuclide is I-123 MIBG/meta Iodo Benzyl guanidine scan.

   Or PET CT scan may also be used.

Staging of Neuroblastoma 00:14:30

INSS/International neuroblastoma staging system is used, according to which there are 4 stages :

Stage 1 : Completely resectable tumors.
Stage 2 : Incompletely resectable tumors.
2a : With no lymph node involvement.
2b : With lymph node involvement (only ipsilateral).

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Stage 3 : Crosses the midline or unilateral tumor with contralateral lymph node involvement.

Stage 4 : Disseminated tumors. widespread metastasis to organs.

Special type : Stage 4S : Stage I or 2 tumor with dissemination to bone/liver/skin & noted in an infant (<1 year).

Stage 4S has good prognosis.

Risk stratification

Low risk

- Stage I
- Stage II
- Stage 4s
- Age of onset <18 months
- Genetics :
  Hyperdiploidy.
  TrK A gene expression.

High risk

- Stage 3/4
- Age >18 months
- Genetics :
  Hypodiploidy.
  N-myc gene amplification.
  Loss of heterozygosity (1p,11q).

Treatment of Neuroblastoma

00:20:54

Treatment

Localised

Surgical resection of tumor

Disseminated

1. Surgery + Chemotherapy.
   Chemotherapeutic agents include :
   Cisplatin.
   Cyclophosphamide.
   Vincristine.
   Etoposide.
2. Radiotherapy is given in cases where surgery and chemotherapy is unsuccessful.
3. Autologous Hematopoeitic stem cell transplant (AHSCT) is used in Refractory cases.

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Wilm's tumor

00:22:30

Solid tumor of the abdomen affects the kidney.
Hence, called as nephroblastoma.
Most common solid tumor of the kidney in childhood.
Second most common intra abdominal tumor in children.

Cell of origin :
Primitive structures in the kidney due to undifferentiated mesenchyme of the kidney called Nephrogenic rests.

Age : 2 to 5 years of age.

Usually unilateral.

Associations/risk factors for development of Wilms tumor :

1. Malformations :
   - Renal malformations like horse shoe kidney confers 4 fold increase risk of Wilms tumor.
   - Cryptorchidism/Undescended testes.
   - Hypospadias.
2. Genetic syndromes :
   - WT-1 gene (11p 13) mutation causes :
     1. WAGR syndrome (Wilm's tumor, Aniridia, Genitourinary anomalies, Growth Retardation).
     2. Denys drash syndrome characterized by diffuse mesangial sclerosis usually presenting as congenital nephrotic syndrome.
        It is associated with ambiguous genitalia in males.
   - WT-2 gene (11p 15) mutation causes :
     Beckwith-Wiedemann syndrome characterized by LGA baby with hemihypertrophy.
     Other symptoms they have are : macroglossia, Omphalocele, organomegaly (liver/spleen) and ear lobe creases.

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Clinical features of Wilm's tumor :

- Asymptomatic abdominal mass in the loin due to enlargement of the kidney.
- Increased production of renin leading to hypertension in children.
- Hematuria.

Other features are related to metastasis :
Lung is the most common site of metastasis.
Followed by lymph node and liver.

Staging :

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Children oncology group (COG) staging is used.
Staging is done after surgery is done.

Stage 1 : Limited or confined to kidney, completely resectable.
Stage 2 : Extends outside the kidney margins, completely resectable.
Lymph nodes are not involved.
Stage 3 : Residual tumor after surgery confined to the abdomen.
Regional lymph node metastasis.
Tumor extension into IVC.
Stage 4 : Lymph node metastasis outside abdomen.
Hematogenous metastasis/metastasis to organs like lung, liver.
Stage 5 : Bilateral kidney involvement.

Treatment :
Nephrectomy is done in all cases.

Nephrectomy

Radical nephrectomy

All other stages

Stages 1/2 : Vincristine with Actinomycin D for 18 weeks.

Stage 3/4 : Vincristine with Actinomycin D and Doxorubicin for 24 weeks.
Radiotherapy.

Partial nephrectomy

Surgery + Chemotherapy + Radiotherapy.

Cases where there are bilateral tumors : Stage 5

Active space

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SHOCK

Shock is the condition of decreased perfusion (decreased O_2) to tissues.

Stages of shock

00:00:33

1. Compensated shock :
   - Restlessness
   - Tachycardia (earliest sign)
   - Tachypnea
   - Capillary refill time (CRT) > 3 seconds : Adequacy of perfusion.

↓

2. Uncompensated shock
   - Drowsiness
   - Oliguria
   - CRT > 5 seconds
   - Hypotension
   - Narrow pulse pressure
   - Cold extremities

↓

3. Irreverible
   - Unresponsiveness
   - Anuria
   - Bradycardia
   - Decreased respiratory rate/ Bradypnea
   - Unrecordable blood pressure

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Types of shock

00:05:28

1. Hypovolemic shock
2. Cardiogenic shock (impaired contractility of heart)
3. Distributive shock :

Active space

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- Blood volume : Normal
- Vasodilation : Pooling of blood (seen in anaphylaxis)

4. Septic shock

Cold	Warm
• Low cardiac output + high SVR	• High cardiac output + low SVR
• Cold extremities	• warm extremities
• Feeble pulses	• Bounding pulses
• Prolonged CRT	• Flash CRT (CRT< 1 second)

Management of shock

00:10:15

1. Early recognition
2. Vigorous treatment :

IV fluids (Crystalloids – NS) :
20 ml/kg over 5 – 10 minutes

↓

60 ml/kg

Assessment : General status
Urine output.
CRT
Heart rate

Fluid refractory shock.

↓

Inotrope :
Initial : Dopamine (10 µg /kg /min upto 15 µg /kg /min)

↓

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↓

Assess the patient

Warm shock	Cold shock
↓	↓
Norepinephrine	Epinephrine

No response

↓

Fluid and Inotrope refractory shock

Active space

↓

Suspect adrenal insufficiency

Hydrocortisone

↓

No response

↓

ECMO (Extracorporeal membrane oxygenation)

In case of cold shock with normal blood pressure, if there is no response to Epinephrine :

↓

- Vasodilators/
- Type 3 PDE inhibitors. Eg : Milrinone

Other measures :

1. Acidosis, pH < 7 : $NaHCO_3$ – 1 mEq/kg.
2. Hypoglycemia.
3. Septic shock : 3rd generation Cephalosporin + Aminoglycoside.

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Active space

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PEDIATRIC ADVANCED LIFE SUPPORT (PALS) GUIDELINES

Given by American Heart Association (AHA) in the year 2020.
Used during Cardiopulmonary arrest (respiratory cause > cardiac cause in children).
Most common causes :

- Respiratory failure.
- Terminal stage of shock.

Overview of CPR :
Assessment ⟶ Circulation ⟶ Airway ⟶ Breathing.

Assessment :
Check for responsiveness by tapping the child.
If the child is unresponsive, check for :

- Circulation by checking the pulse.
- Breathing by checking chest movements.

Assessment should be done within 10 seconds.
Circulation and breathing are assessed to confirm if the child is in cardiopulmonary arrest.

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Circulation :
By starting chest compressions using the techniques :

- Two thumb technique : The child's chest is encircled with both the hands and two thumbs are pressed over the lower 1/3rd of the sternum.
  Used in children < 1 year.
- Chest compressions are given with one hand by placing the base/heel of the hand over the lower 1/3rd of the sternum.
  Used in children between 1-8 years.
- Chest compressions are given using both the hands b interlocking them and placing the base/heel of the hands over the lower 1/3rd of the sternum.
  For children > 8years.

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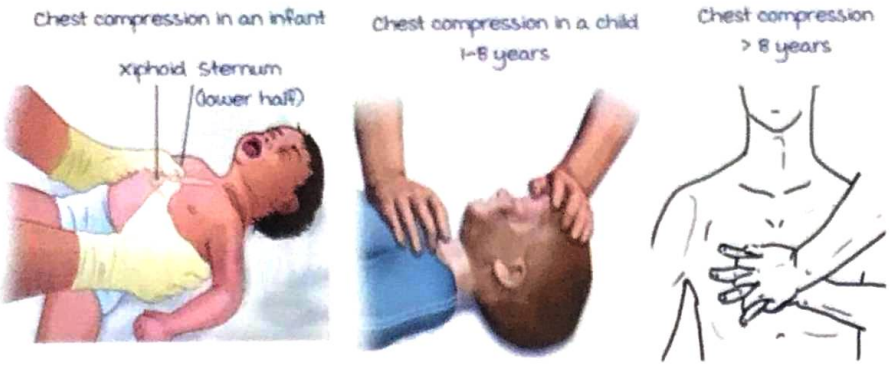
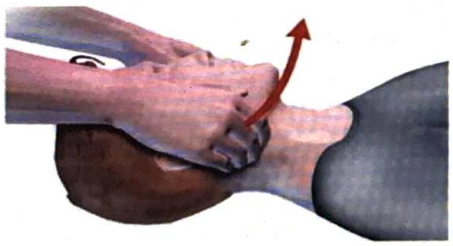


Push hard and push fast principle should be followed while giving chest compressions. Allow full recoil before next push.
Rate : 100-120/minute.
Depth : 1/3rd of the AP diameter of the chest.
Allow full recoil of the chest in between the compressions.
Site : Lower 1/3rd of the sternum. Avoid the xiphoid process.

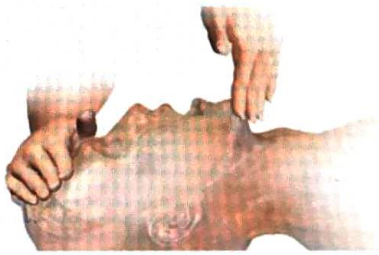
Airway

00:06:48

- Jaw thrust : Fingers of both the hands are kept behind the angle of the mandible and the jaw is lifted upwards and slightly outwards.
- Head tilt and chin lift : One hand is used to push the forehead back and the other hand is used to lift the chin. 60c6b3eeaa8ded0e4e7e5ea7
  This manoeuvre is avoided in head and neck trauma because of the possibility of a cervical spinal cord injury.



Jaw thrust



Head tilt, chin lift

Breathing

00:08:54

Types of manual breathing/assisted ventilation :

- Non-invasive form : Bag and mask ventilation.
- Invasive form : Endotracheal intubation.

Size of the ET tube : (Age of child in years/4) + 4.

Active space

Leave Feedba

Type of the ET tube : Cuffed ET tube.
It prevents air leaks and decreases the number of times the tube has to be changed.
Cuff pressure : < 20–25 cm H_2O.
Higher pressures can lead to pressure induced tracheal necrosis.

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Depth of insertion of ET :

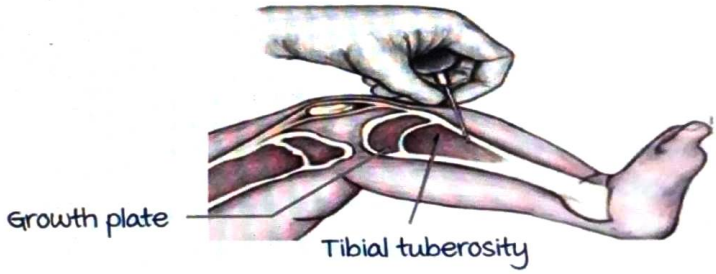
- 3 x size of the ET (in cm).
- [Age (years)/2] + 12.

Newer updates of PALS 2020 guidelines :

1. Routine use of cricoid pressure during endotracheal intubation is not recommended.
2. Rate of ventilation : 20–30 breaths/minute i.e., 1 breath every 2–3 seconds.
3. Naloxone should not be used routinely in all babies with respiratory depression.
   It is used only in cases of opioid overdose.
4. In out of hospital cardiopulmonary arrest, bag and mask ventilation is as good as endotracheal intubation.

Vascular access

00:14:13



Pediatric intraosseous access

For administration of medications like Epinephrine.
Recommended route :

- Intravascular access (i.v route).
- Large accessible vein (most common : Femoral vein).

Alternatives : Intraosseous access (IO) :

The needle is inserted below and medial to the tibial tuberosity.
Proximal 1–2cm of tibia should be left undisturbed due to the presence of the growth plate.

Active space

Algorithm (protocol) of CPR

00:16:28

Unresponsive patient

↓

Pulse/breathing (≤ 10 seconds)

↓

Pulse absent
Breathing absent

↓

Start high quality CPR
Compression + ventilation
Single rescuer (30 : 2)
Two rescuer (15 : 2)

↓

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Reassess after 2 minutes /2 cycles

↓

Attach AED

↓

Assess the rhythm

↓

Shockable rhythm (PVT, VF)

↓

Shock : 2 J/kg
Continue CPR

↓

Reasses after 2 minutes

↓

No response

↓

Repeat shock
max 10 J/kg

Non shockable rhythm (Asystole, PEA)

↓

Inj Epinephrine IV
0.1 ml/kg (1 : 10000)

↓

Continue CPR
Reassess after 2 minutes & take further decision

Pulse present
Breathing absent

↓

Start rescue breathing
20-30 breaths/minute

↓

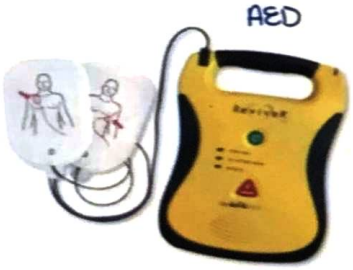
Reassess every 2 minutes

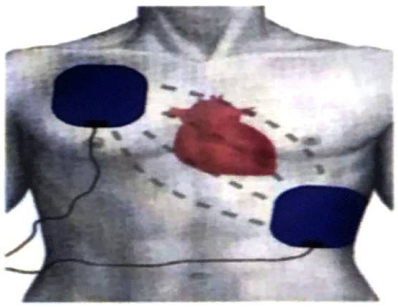
↓

Pulse absent
or HR< 60 bpm with signs of poor perfusion

→ Start high quality CPR

AED





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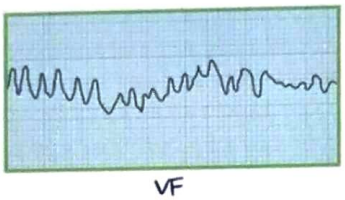
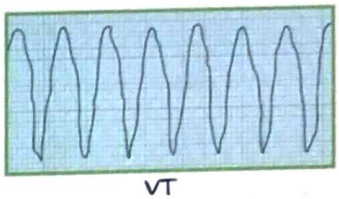
AED (Automated Electrical Defibrillator) :
Has two pads which are attached over the precordium.
It displays the rhythm after it has been attached.

Shockable rhythms :

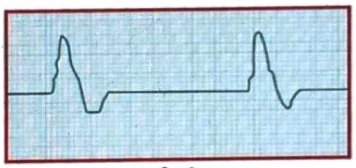
- VF/Ventricular Fibrillation : Irregular rhythm with broad QRS complexes.
- VT/Ventricular Tachycardia : Regular rhythm with broad QRS complexes. Also known as Pulseless VT (PVT).

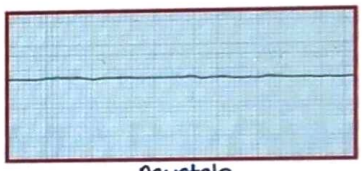

VF


VT

Non Shockable rhythms :

- Asystole : Flat line.
- PEA/Pulseless Electrical Activity : Occasional electrical activity without any heart rate.
- Administer epinephrine as early as possible (better outcome).


PEA


Asystole

Reversible causes of cardiac arrest :

4 H	4 T
Hypovolemia	Tension pneumothorax
Hypoxia	Cardiac Tamponade
Hypothermia	Toxins/drugs
Hyperkalemia	Thromboembolism (Pulmonary)

Post resuscitation care

00:28:15

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active space

After the return of spontaneous circulation following successful CPR, the following are to be corrected and monitored :

- Maintenance of BP.
- Correction of hypoxia.
- Correction of hypo/hypercapnia.

- Targeted temperature management (decreasing the body temperature to 36–37.5 °C because it reduces the metabolic activity in the brain and leads to its recovery if there was any brain damage that had occurred during cardiac arrest).
- Continuous EEG monitoring (to detect non convulsive seizures).
- Delaying the prognosis for 72 hours.

Summary 00:31:04

- High quality cardiopulmonary resuscitation (CPR) is the foundation of resuscitation (push hard, push fast).
- A respiratory rate of 20 to 30 breaths per minute to be provided.
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- For patients with nonshockable rhythms, the earlier epinephrine is administered, better the outcome.
- Use a cuffed endotracheal tube.
- The routine use of cricoid pressure is not indicated.
- In out of hospital cardiopulmonary arrest, Bag and mask ventilation is as good as endotracheal intubation.
- Excellent post cardiac arrest care is critically important.

PALS Guidelines : Bradycardia Algorithm 00:32:08

Paediatric bradycardia : Heart rate less than 60 per minute.

Basics:

1. Look / Treat the cause : conditions like hypoxia, hypothermia & medications.
2. Maintain a patent airway.
3. Oxygen support only when necessary.
4. Connect a cardiac monitor and continuously monitor the vital parameters.
5. Intravenous / Intraosseous access.
6. 12 lead ECG.

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Check for signs of poor perfusion :

- Altered mental status.
- Low BP.
- Shock.

No signs : Stable, close observation.

Signs present :

- Secure airway
- Provide O_2 } Oxygenation

↓

No improvement, HR < 60/min

↓

Provide high quality CPR at lower end of sternum avoiding the xiphoid process — Start CPR

↓ Remains in bradycardia

medications

Medications :

- Epinephrine.

Dose is 0.1 ml/kg (= 0.01 mg/kg).

Strength is 1 : 10,000. Repeat after 3 to 5 minutes.

- Atropine.

Enhances AV conduction, used in Primary AV blocks / High Vagal tone.

The dose is 0.02mg/kg.

The minimum dose is 0.1 mg, maximum is 0.5 mg.

If no response ⟶ Repeat one more dose.

↓ No response to Atropine — AV block

Pacing – Transthoracic/ Transvenous.

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PALS Guidelines : Tachycardia Algorithm 00:40:28

Tachycardia

↓

- Support ABC / Airway, Breathing, Circulation.
- Provide O2.
- Attach cardiac monitor/defibrillator.

↓ Tachycardia persists

12 lead ECG

↓

QRS duration

QRS duration : Wide : > 0.08 sec

Narrow : < 0.08 sec

Narrow complex QRS:

1. Sinus tachycardia	2. PSVT / Paroxysmal Supraventricular Tachycardia.
• Consistent history. • HR < 180/min : Children. < 220/min : Infants. • Variable R-R interval. • Constant P-R interval. • P wave is present. • Treat cause (fever, Hypoxia, Hypoglycaemia.)	• Abrupt onset / Inconsistent history. • HR > 180/min : Children. > 220/min : Infants. • Fixed R- R interval. • P waves are Absent / Abnormal.

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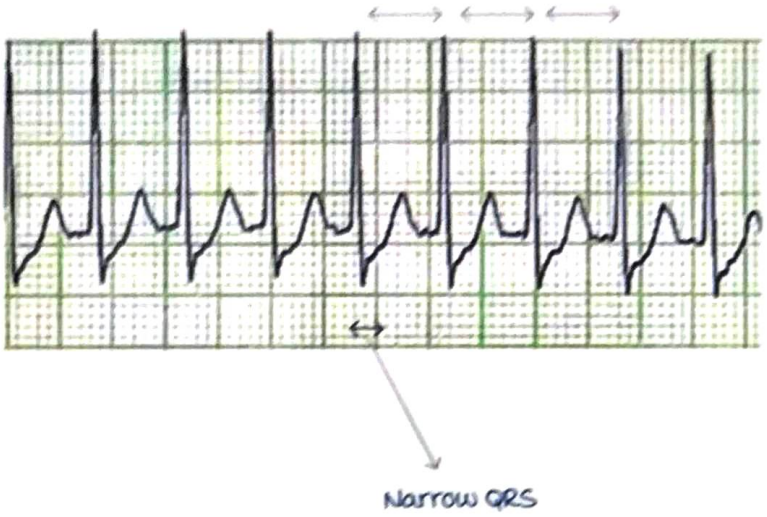
PSVT :



Narrow QRS : 0.08 sec.

P wave is absent.

R-R interval is constant.

Treatment of PSVT:

Consider vagal manoeuvres : It decreases the rate of conduction thereby decreasing the heart rate.

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Ice pack to upper half of face avoiding nose and mouth.

Do not apply excess ocular pressure (can lead to retinal damage).

Older child : Valsalva manoeuvre.

Drugs:

Drug of choice : I.V. Adenosine 0.1mg/kg (max : 6mg).

Rapid bolus technique (short half-life of adenosine) :

with two syringes .

(Adenosine quickly by 1st syringe & 3 to 5ml of normal saline by 2nd syringe)

↓ No response.

Adenosine I.V. 0.2mg/kg (max – 12mg) again by rapid bolus technique.

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If not responding to Adenosine or I.V access is not available,

Next step : Synchronised cardioversion.

0.5J/kg → 1J /kg → 2J/kg (maximum dose).

Wide complex QRS (>0.08 sec) tachycardia :

Usually Ventricular tachycardia : Shockable rhythms.

Treatment of choice : Synchronised cardioversion.

0.5J/kg → 1J /kg → 2J/kg (maximum dose).

↓ No response

Drugs :

Amiodarone : 5mg/kg i.v

Over 20 to 60 minutes.

Procainamide : 15mg/kg I.V or I.O

Over 20 to 30 minutes.

With refractory ventricular tachycardia,

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Cardiologist opinion is recommended.

Do not combine Amiodarone & Procainamide.

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