Pediatrics

NEONATOLOGY

LBW- <2.5kg
VLBW- <1.5kg
ELBW- <1kg
SGA- <10th
AGA- 10th - 90th percentile
LGA- >90th → BWS / IDDM / Sotos Sx

Physiological weight loss:
term – 10% – regain -d10
preterm – 15% – regain - d15

IUGR: efw <10th percentile / Doppler abn

	Symmetrical	Asymmetrical
Etiology	Cong anomaly / infx	UPI
HC	↓	"brain sparing"
AC	↓	↓
Ponderal index g/cm³	>2 (N)	<2
Prognosis	poor	good

MCC Of mortality: RDS / IVH / NEC
Neonate – Prematurity > Asphyxia > Sepsis > Congenital anomalies (PAS-C)
U5- Prematurity > Neonatal infection> Asphyxia > Pneumonia > Diarrhea

Expanded new Ballard score: 20 - 44 wks

Neuromuscular Maturity

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

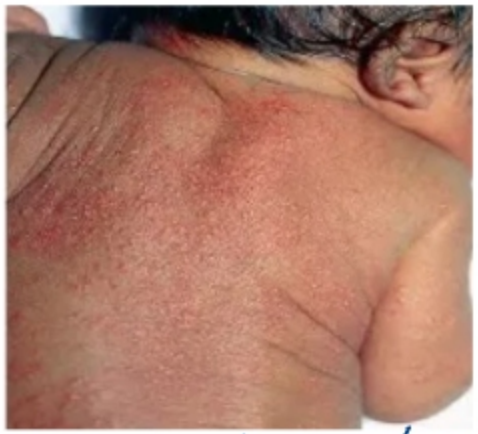
Physical Maturity

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	

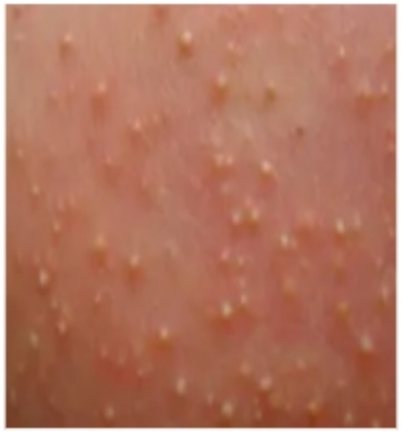
Maturity Rating

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

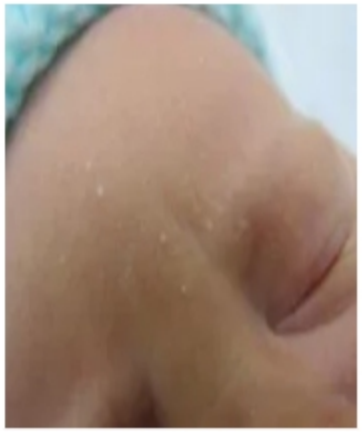
Benign newborn lesions



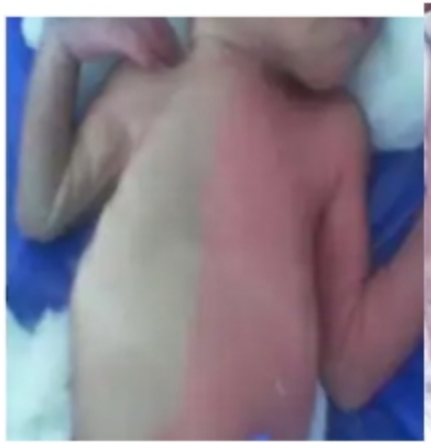
Erythema toxicum / neonatorum
(eosinophils ↑



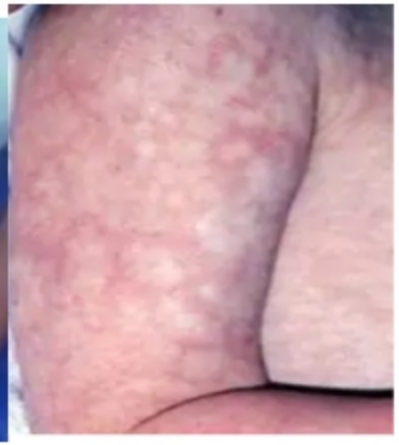
Neonatal
(pustulosis
↑neutrophils



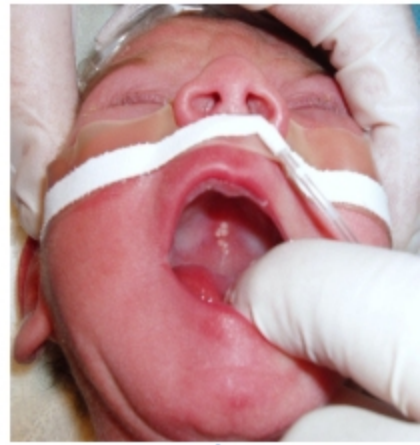
Milia
(epidermal inclusion
cyst



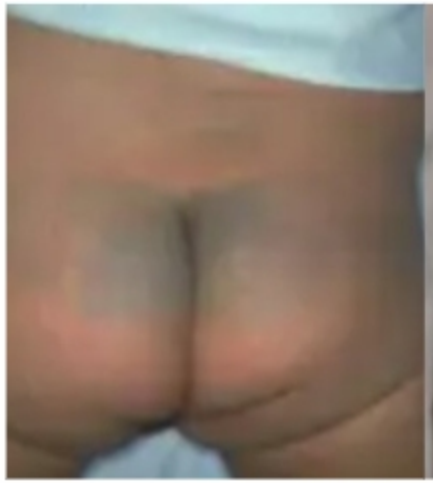
Harlequin
change



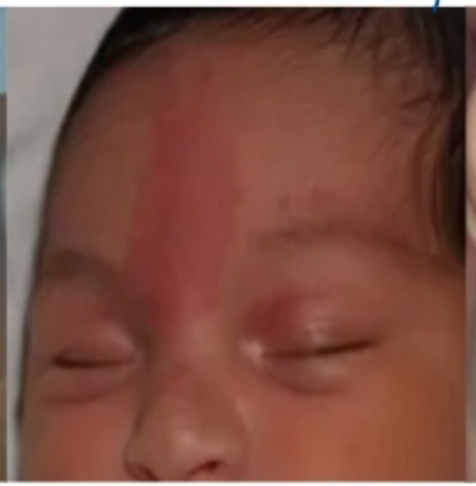
Cutis marmorata



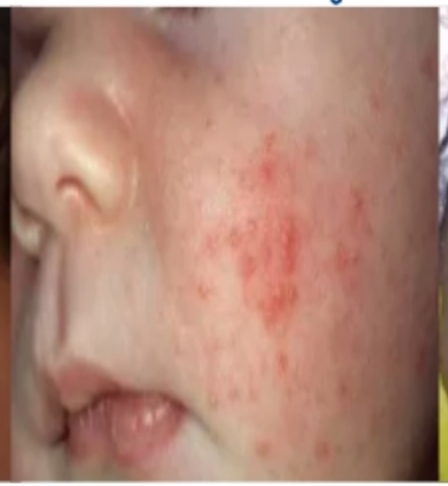
Epstein
pearls
(keratin)



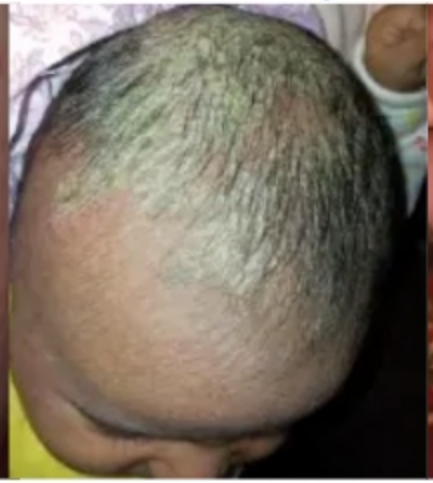
Mongolian spots



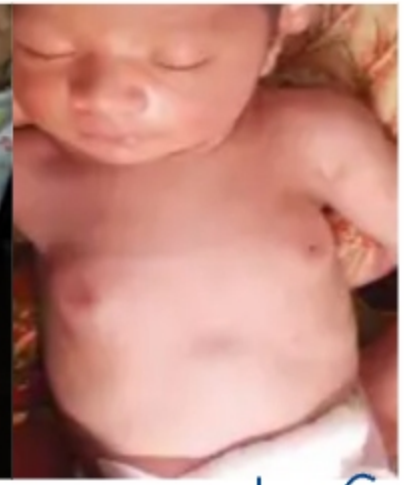
Strawberry Hemangioma /
stork bite /
infantile Hemangioma
(involutes by 1-2yrs.



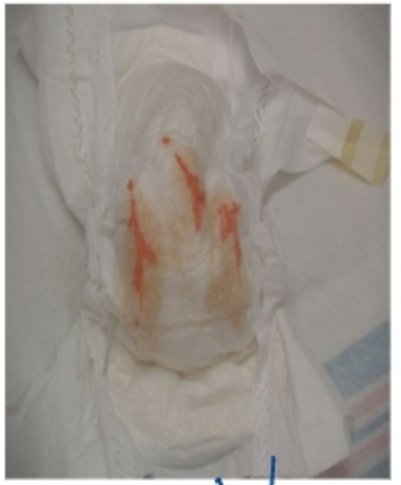
Acne



Cradle cap /
Seborrheic Dermatitis



↓ (mat Estrogen ↑) ↓
mastitis
neonatorum



pseudo-
menemachia

HYPOTHERMIA

37.5° **Normal range**

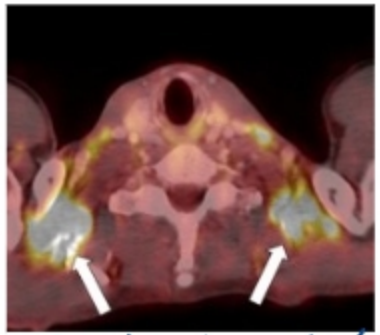
36.5° **Cold stress**

36.0° **Moderate hypothermia**

32.0° **Severe hypothermia**

temp → axilla x 3min

Non shivering thermogenesis
- Brown fat – nape of neck / interscapular
- False +ve: PET



Retained brown fat / Hibernoma

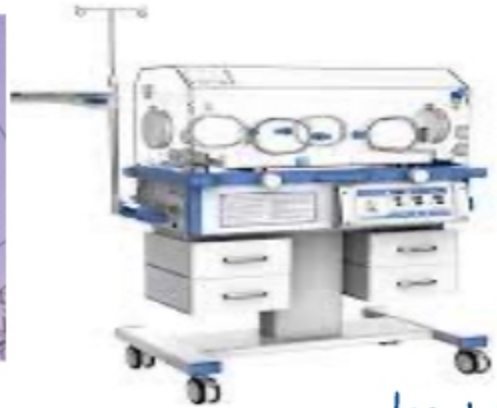
heat loss

KMC

1. Kangaroo position (skin to skin) → prevent hypothermia (conduction)
2. Kangaroo nutrition → Exclusive bf
3. Kangaroo support
4. Kangaroo discharge - early - ↓ n. sepsis

Indication: All stable LBW

min ~1hr - as long as possible



Incubator =) Convection

Radiant warmer (Radiation

Neonatal Hypoglycemia

Definition: Blood <40mg/dl / Plasma <45mg/dl

Asymptomatic:
- [IDM, preterm, sick]
- >20mg/dl → oral (BF) → >40mg/dl - continue
  - <40mg/dl
- <20mg/dl → glc infusion

Symptomatic: iv dextrose bolus 2ml/kg → infusion
jitteriness, sz, coma

Features Differentiating Jitteriness from Seizures

1. Absence of eye deviation or fixed gaze, heart rate changes.
2. Rhythmic tremors (7-10Hz) with equal to and fro movements VS seizure is slower (1–2 Hz), with rapid and slow components.
3. Stimulus sensitive, precipitated by hunger, crying, or loud noise, and stopped by gentle restraint.

Neonatal sepsis

- -MCC of neonatal sepsis- Acinetobacter > Klebsiella
- -MCC of early onset sepsis (<72hrs)- Grp B Strep
- -Most effective method for prevention- Handwashing
- -Earliest C/F: inad feeding
- -Initial Ix: Sepsis screen -IOC: Blood culture
- -R/F: Preterm, ROM >18hrs, PPV
- Rx: Empirical: Ampicillin + Gentamicin ± Vancomycin (after blood culture)

Sepsis Screen ≥ 2/5 +ve

- Leukopenia (TLC < 5000)
- Neutropenia (ANC < 1800)
- Immature to total neutrophil (I/T) ratio >0.2
- Micro-ESR > 15mm 1st hour
- CRP + ve

Normal Urine: ≤ 24 hrs
Meconium: ≤ 48 hrs
(biliverdin)

NEONATAL JAUNDICE

PATHOLOGICAL:

- Appears <24hrs

Causes: ABO incompatability (O-mother) / G6PD / HS

- Jaundice persisting > 14 days = Prolonged

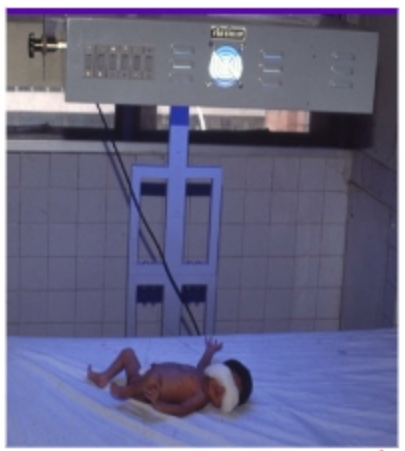
Causes: Hypothyroid / Cephalhematoma / breast milk jaundice

- Increase of bilirubin >5mg/dl/day
- Serum bilirubin>15mg/dl
- Conjugated Bilirubin >1mg/dl

Kramer's

Area of Body	Approximate Serum Bilirubin Level
Face	4–6 mg/dL
Chest	8–10 mg/dL
Abdomen	10-12 mg/dL
Limbs	12-14 mg/dL
Palms, soles	>15 mg/dL

↳ TCB +ve → total serum bilirubin



SE: Bronze baby / gonadal / retinal / diarrhea

Mechanism:

Structural isomerization: most imp – Bil → Lumirubin

Photo isomerization: Z → E

Photo-oxidation least imp

-Distance: 30-45cm

-Wavelength: 450nm

-Type of lamp: LED

-Irradiance using flux meter:30 uW/cm2/nm

	Phototherapy	Exchange transfusion
24-48hrs	>15	>20
48-72hrs	>18	>25
>72hrs	>20	>25

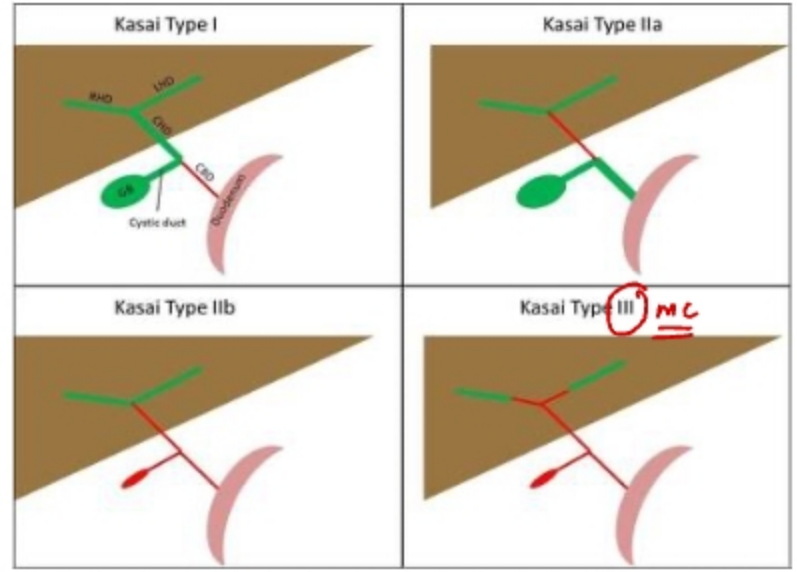
Lille's chart

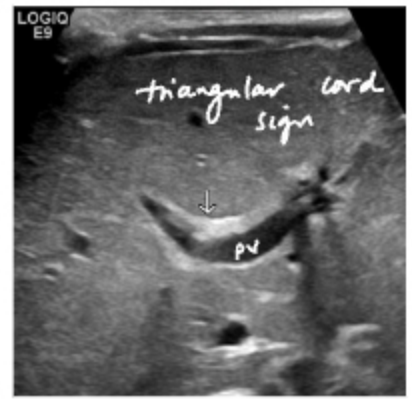
UNCONJUGATED

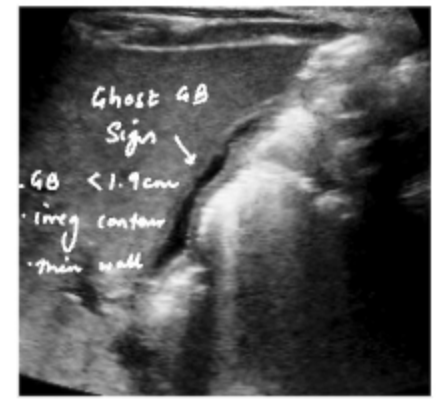
Crigler Najjar AR	Gilbert AR	BREAST MILK JAUNDICE	BREASTFEEDING JAUNDICE
UDP-GT severely deficient type 1 absent type 2 ↓↓ Phenobarbitone × improve improve	UDP-GT mildly deficient	Prolonged jaundice pregnanediol / ffa ↳ UDP-GT ⊖ - 3% babies - bf ≥ - Rarely phototherapy - STOP Bf	First week - Inadeq feeding (↑↑ enterohep circuit

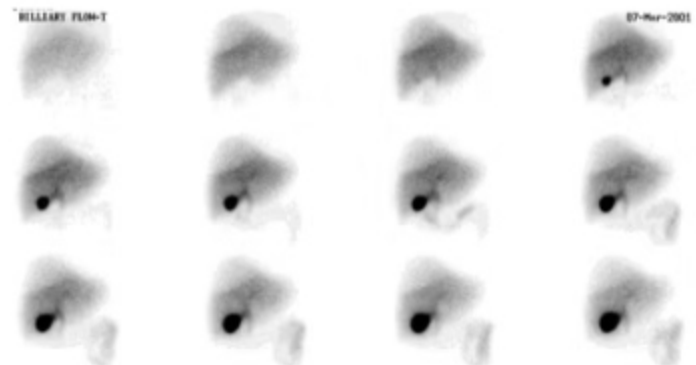
CONJUGATED: Direct bilirubin >2mg/dl

Dubin - Johnson AR	Rotor AR	EHBA	Neonatal hepatitis
MRP-2 Black liver epiN metabolites	No black liver	Periductal fibrosis and proliferation IOC: fasting USG > Tc^{99}-HIDA > Liver biopsy > Intra-op cholangiography TOC: Kasai procedure ↳ Good / × cirrhosis ↓ × Liver transplant	Giant cells, lymphocytes









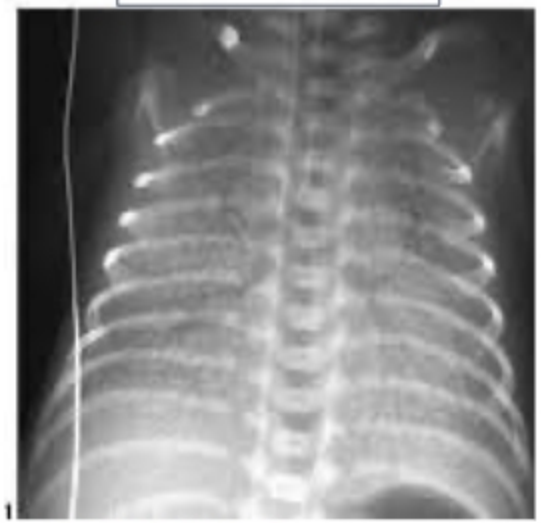
Tc^{99m} - HIDA scan
(N)

EHBA → Non-visualised GB
(↑ NPV)

Neonatal respiratory distress

RDS / HMD

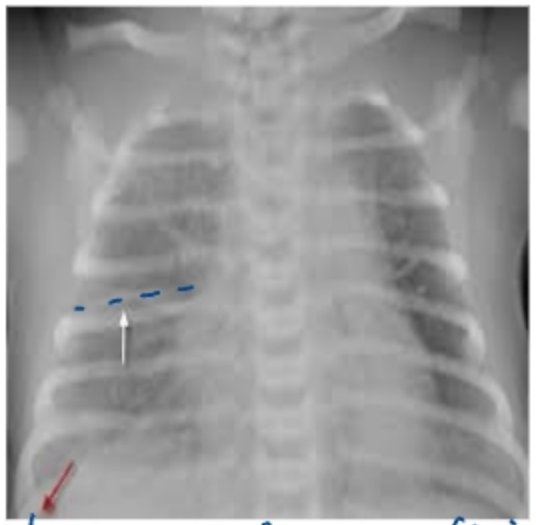
PRETERM / IODM



- Underinflated
- White out lungs
- air bronchogram

TTNB

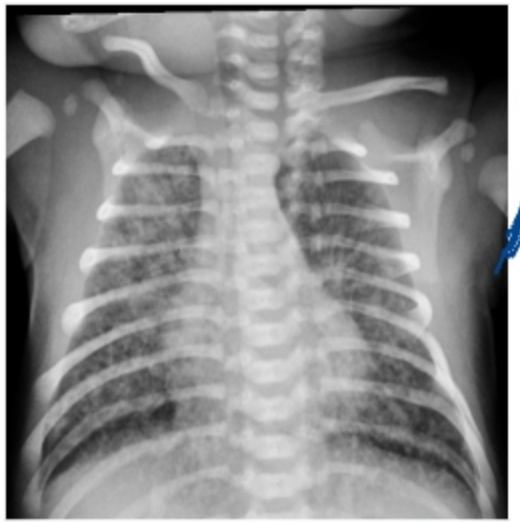
TERM / LSCS



↳ CPL / fissure fluid
↳ ENaC → absorpn of fluid

MAS

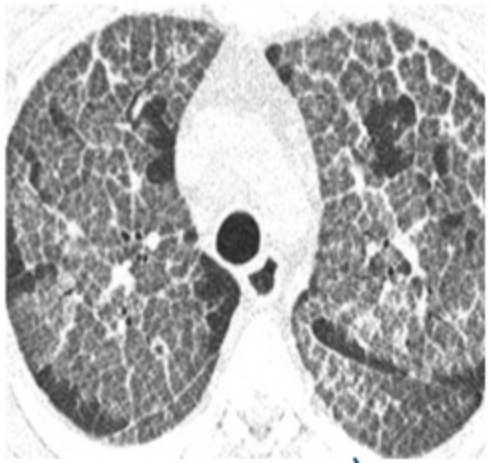
POST-TERM MSL



Hyperinflation / Steaky opacities

PAP – Pulm alveolar proteinosis

H/o sibling death
SP-B – mphges



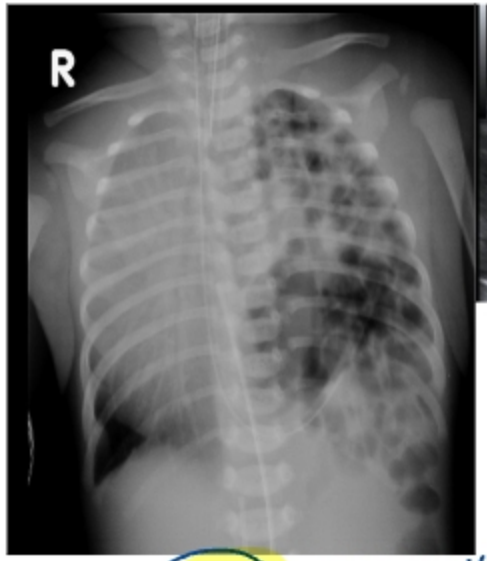
Crazy paving
Rx- whole lung lavage

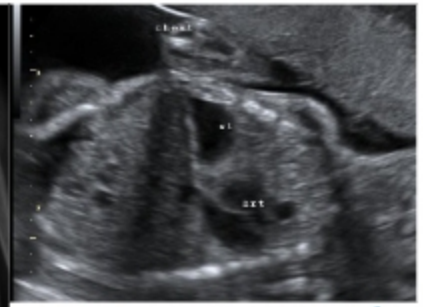
Surfactant- Type 2 pneumocytes
Synthesis begins- 20 wks
Amniotic fluid- 28 wks
Mature lungs- 34 wks
Lecithin: Sphingomyelin ratio > 2:1 (N)
(Dipalmitoyl phosphatidylcholine)
Nile blue sulfatase test - orange – mature lungs

Shake test (+ 95% ethanol)

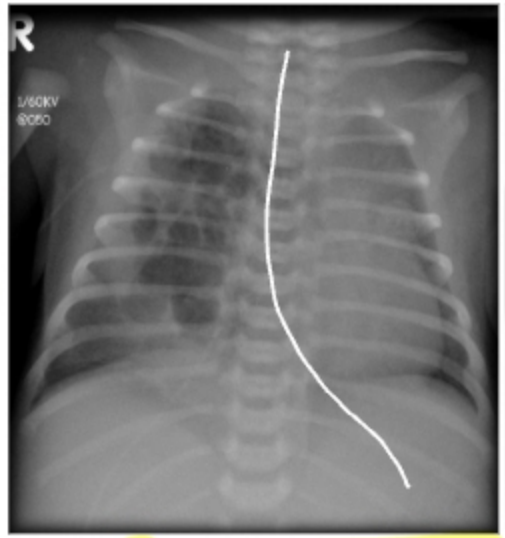
Rx of HMD: CPAP / Mechanical ventin – fiO_2 ≥ 40% → Surfactant
INSURE: Intubate → surf → extubate asap
LISA / MIST: Less inv surf admin Rx / Min inv surf Rx
Prophylactic surfactant: ≤ 28 wks POG
Failure to "wean off": Bronchopulm dysplasia "cystic lucencies"
1-7 days Preterm: Apnea of prematurity → ≥ 20s + cyanosis / bradycardia
Rx: Caffeine

Congenital pulmonary malformations

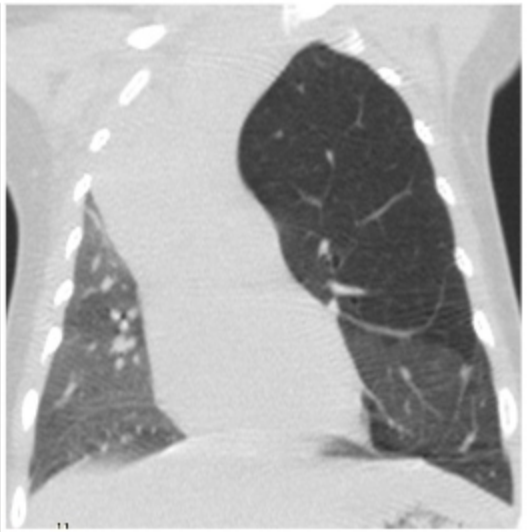




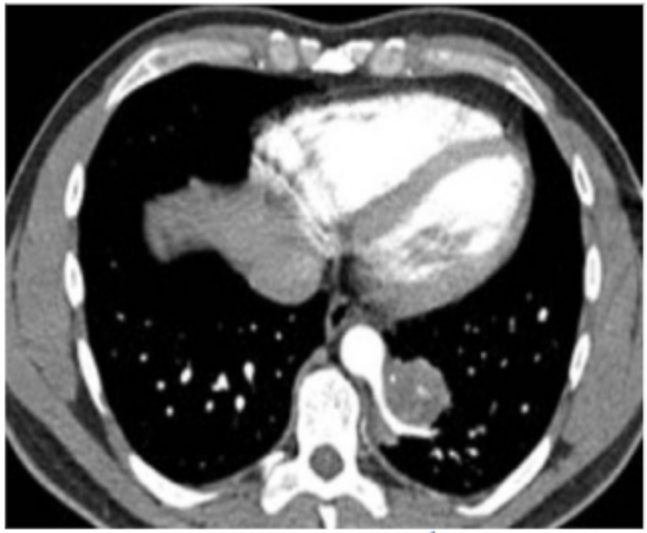
CDH - antenatal USG
↓
prognostic - LHR
Lung : head ratio
<1 - ☹
↓
EXIT procedure
ex-utero
Intrapartum



CPAM = cystic pulm airway malform"



CLO / CLH
Cong lobar
overinfl" / hyperinflation
MC → LUL

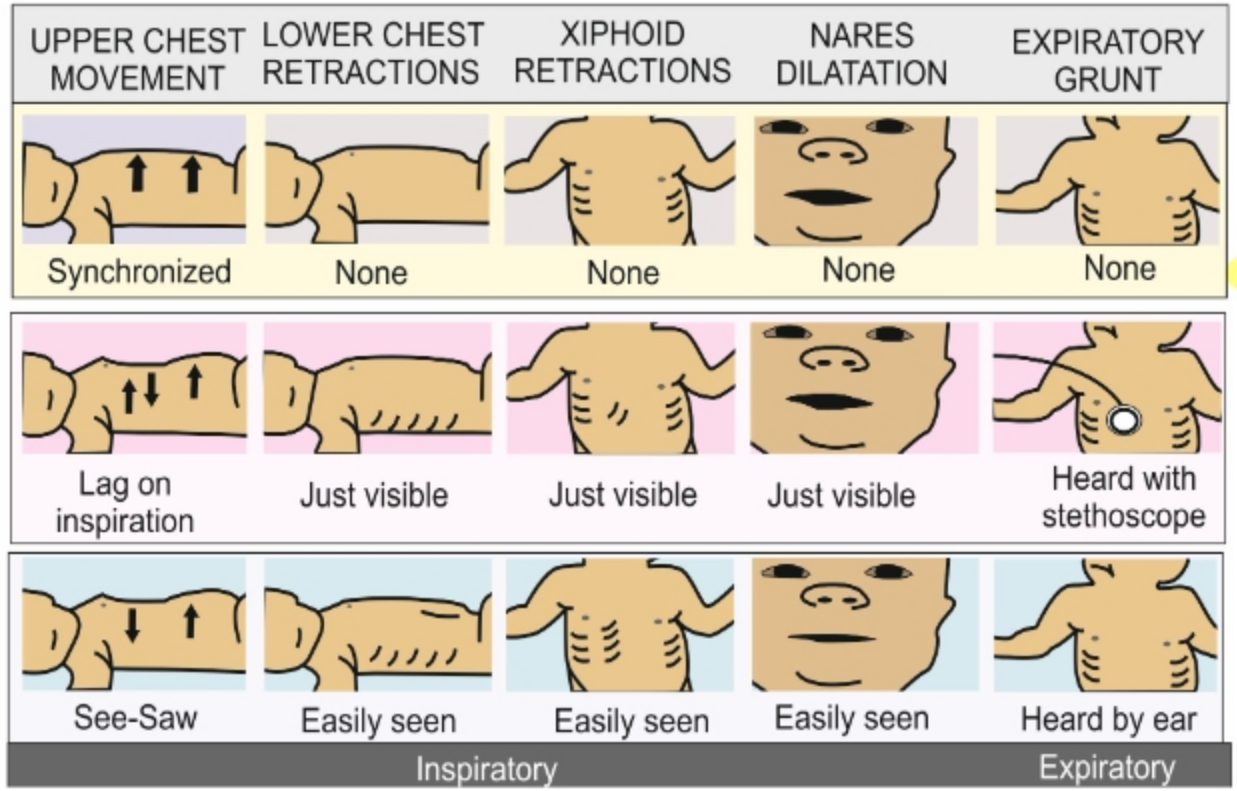


Sequestration
↳ Aorta - branches
extra-lobar
- systemic venous
intra-lobar
- p. vein
- MC → LLL
- recurrent pneumonia

CDH - pleuro peritoneal membrane ✗
- Morgagni
  - ant / Rt
  - adults
  - incidental
- Bochdalek (mc)
  'BPL'
  post / Lt
  ↓
  neonatal resp distress
  "scaphoid abdomen"
  → BMV CI

Most imp progn - Pulm hypoplasia aa

Ist - ET tube → NG tube
- CXR
- Sx correction



Silverman -Anderson -Preterm

Score	0	1	2
Respirate Rate	<60	60-80	>80
Cyanosis	None	No cyanosis with oxygen	Cyanosis with oxygen
Retraction	None	Mild	Moderate to severe
Grunting	None	Audible with stethoscope	Audible without stethoscope
Air Entry	Good	Decreased	Barely Audible

Downe's - term / preterm

APGAR

APGAR Score	Score 2	Score 1	Score 0
Appearance	 Pink	 Extremities Blue	 Pale or Blue
Pulse	>100 bpm	<100 bpm	No pulse
Grimace	Cries and pulls away	Grimaces or weak cry	No response to stimulation
Activity	 Active movement	 Arms, legs flexed	 No movement
Resp effort	Strong cry	Slow, irregular	No breathing

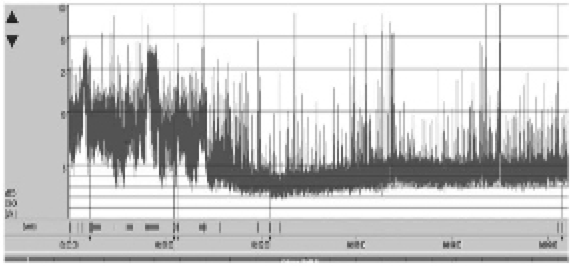
Prognostic score
Timing: 1, 5 min

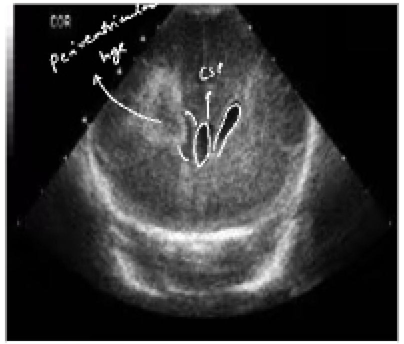
Severe asphyxia:
- **APGAR:** < 3 at 5 min
- **pH:** < 7.0
- **CNS dysfunction:**
- **Hypotonia/seizures/ AbN Moro**
- **MODS**

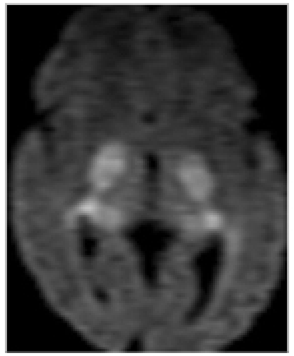
HIE

Neonatal seizures
MC TYPE- Subtle (immature cortical network)
Best prognosis- Focal clonic
Worst prognosis- Myoclonic
MC CAUSE- HIE
CAUSES- ↓glycemia / ↓/↑Na / ↓Ca / ↓Mg
DOC- Phenobarbitone (↑GABA) —×→ Pyridoxine (B6) Glu ↓ GABA

BEDSIDE MONITOR- Integrated amplitude EEG
INITIAL IX- transcranial USG
IOC- DWI -MRI

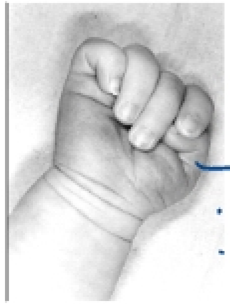






Patterns:
Cerebral palsy-SPASTIC type MC
Non-reversible, non-progressive
Term infant
- Parasagittal injury: Spastic quadriplegia
- Status marmoratus: Choreo-athetoid (BG) - kernicterus

Preterm infant
- Periventricular leukomalacia Spastic diplegia



primitive reflex persistent
- cortical thumb
- Commando crawl
- scissoring gait

Sarnat & Sarnat

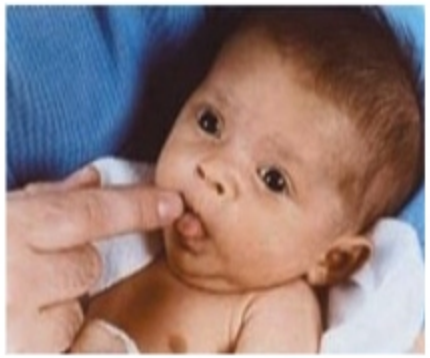
Parameter	Stage 1 (Mild)	Stage 2 (Moderate)	Stage 3 (Severe)
Consciousness	Hyperalert	Lethargic	Coma
Activity	Normal	Decreased	Absent
Moro	Strong	Weak	Absent
Pupils	Mydriasis	Miosis	Variable
Heart rate	Tachycardia	Bradycardia	Variable
Seizures S2	None	Common	Uncommon
Prognosis	99% normal	80% normal	50% death 50% sequelae

Neonatal reflexes

Palmar grasp

A : 28wks

D : 3mon

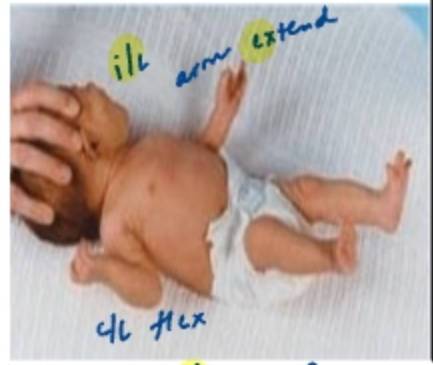


Rooting reflex

A : 32wks

D : 1mon

(1st to disappear)

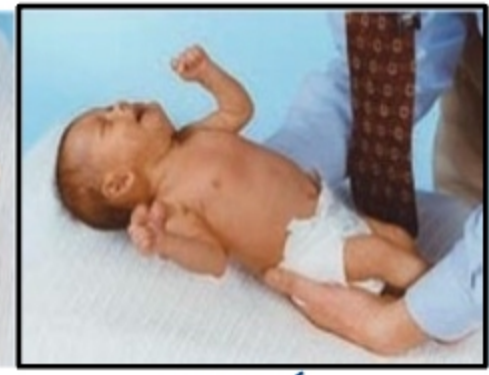


ASTNR

A: 35wks

D: 5-6mon

↳ "Rolling"



MORO'S

- Hands open
- Shoulder abduct /extend/ ant flexion arm

A: 28 - 37wks

D: 6mon

Parachute reflex

A: ~8mon PN

never disappear

Landau reflex

A: 3-4mon PN

D: 2yr

Persistent Moro's: Cerebral palsy
Exaggerated Moro's : HIE st 1
Absent Moro's: HIE st 2/3, Down's, kernicterus
Asymmetric Moro's: clavicle # /Erb's / humerus #

Appear after birth:

. STNR - i/L arm flexed

A: 6-9mon PN

D: 12mon

↳ "creeping / crawling"

Parachute

· Landau

Neonatal Feeding

Gestational age	Maturation of feeding skills	Initial feeding skills
<28 weeks	Inadequate sucking efforts Lack of gut motility	IV fluids
28-31 weeks	Sucking burst develop Lack of coordination between suck, swallow and breathing	Orogastric or nasogastric feeding
32-34 weeks	Coordination between breathing and swallowing begins	Spoon feeding
>34 weeks	Mature sucking pattern	Breastfeeding

'Garage feeding'

Paladi

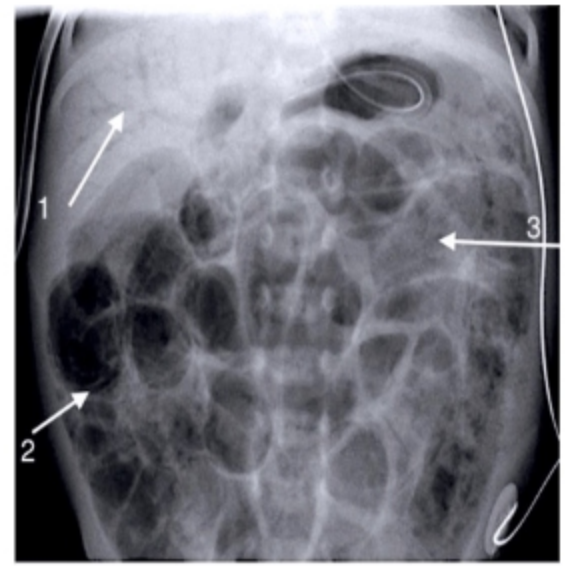
Neonatal fluid requirement:
<1500g: 80ml/kg
>1500g: 60ml/kg
>1week: 150ml /kg

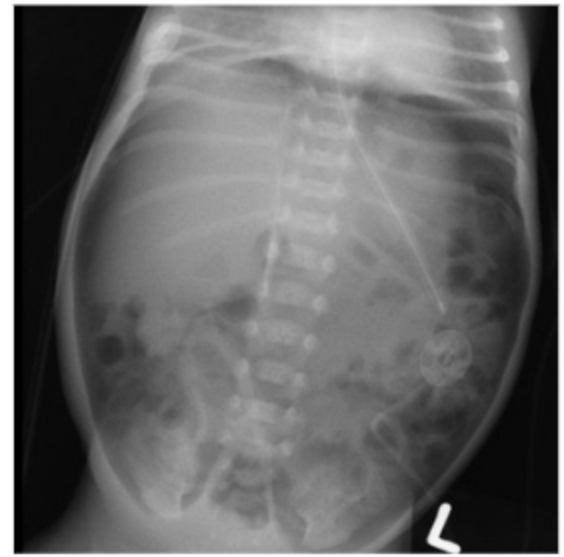
NEC

R/F: Preterm, Formula, PDA

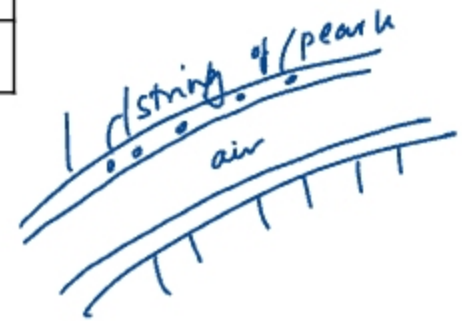
Mod. Bell's

Stage	Systemic Signs	Treatment
IA BAT	Bradycardia, Apnea, Temperature instability	NPO Antibiotics Fluid resuscitation
IB	Grossly bloody stool	
IIA	Absent bowel sounds Pneumatosis intestinalis	
IIB	Metabolic acidosis, Thrombocytopenia PV gas	
IIIA	DIC	
IIIB	Pneumoperitoneum	Surgery





Foot ball sign



TORCH infections

IgG ++
IgM =/= / PCR =/=

Toxoplasmosis — cat ++

- Hydrocephalus
- Chorioretinitis - Headlight in fog / macular scar
- Parenchymal calcification
- Max transmission in T3 Q

CMV QQ

- Microcephaly HC <-2SD / 3rd centile
- Periventricular calcification
- MC long-term sequelae: SNHL QQ
- Most are asymptomatic (90%)
- Urinary PCR test - CMV
- Max r/o transmission from primary CMV

Zika virus (Aedes)

- Microcephaly
- Contractures
- GM-WM calcification
- TIM1 and TAM-XL Q

Varicella (congenital)

- Cicatricial skin rash
- Limb hypoplasia
- Max: 13-20 weeks Q/

VZIG to pregn female (non immune)

- Neonatal varicella syndrome:
- VZIG to infant: 5d before/ 2d after delivery

HSV-2 (active ulcers → LSCS =/=)

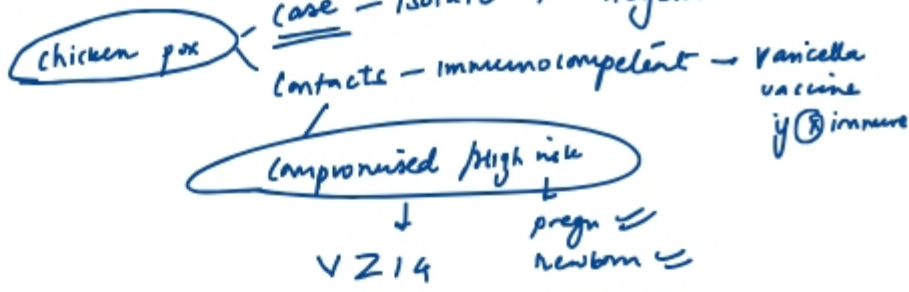
- Skin and eye lesions
- Encephalitis

Congenital rubella

- SNHL-MC finding
- Cataract → Nuclear pearly > Lamellar / zonular
- MC eye C/F: salt-pepper retinopathy
- PDA > PS
- Expanded rubella: DM, renal diseases
- Max transmission in in 1st trimester
- Least r/o perinatal transmission Q

Parvovirus B19

- Non-immune hydrops
- PRCA


chicken pox
case – isolate + Acyclovir
contacts – immunocompetent – varicella vaccine if immune
compromised / high risk
VZIG
pregn
newborn

Growth

Weight with age:
- **Birth** – 2x
- **20-40g/day x 3months**
- **400g/month till 1 year**
- **5mon** – 2x
- **1yr** – 3x
- **2yr** – 4x
- **3yr** – 5x
- **5yr** – 6x
- **7yr** – 7x
- **10yr** – 10x

Height with age:
- **Birth** – 50cm
- **3mon** – 60cm
- **1yr** – 75cm (50% ↑)
- **2yr** – 90cm (½ adult)
- **4yr** – 100cm
- **6cm / yr till 12yrs (150cm)**

US:LS : Pubic symphysis
- **Birth** 1.8 : 1
- **3yr** 1.3 : 1
- **7yr** 1 : 1
- **>10yrs: 0.9:1**

HC
- **Birth-32-35cm**
- **1st 3month:** 2cm / month
- **Next 3month:** 1cm / mon
- **Next 6month:** 0.5cm / mon
- **Next 2 yrs:** 0.2cm / mon
- **>2cm/month abN**
- **12yrs: 52cm**

Shaker's tape

MUAC + age independent
- 6m – 5yr

35cm
+ 6
+ 3
+ 3
] 12cm
= 47cm


Infantometer ≤ 2yrs

Stadiometer

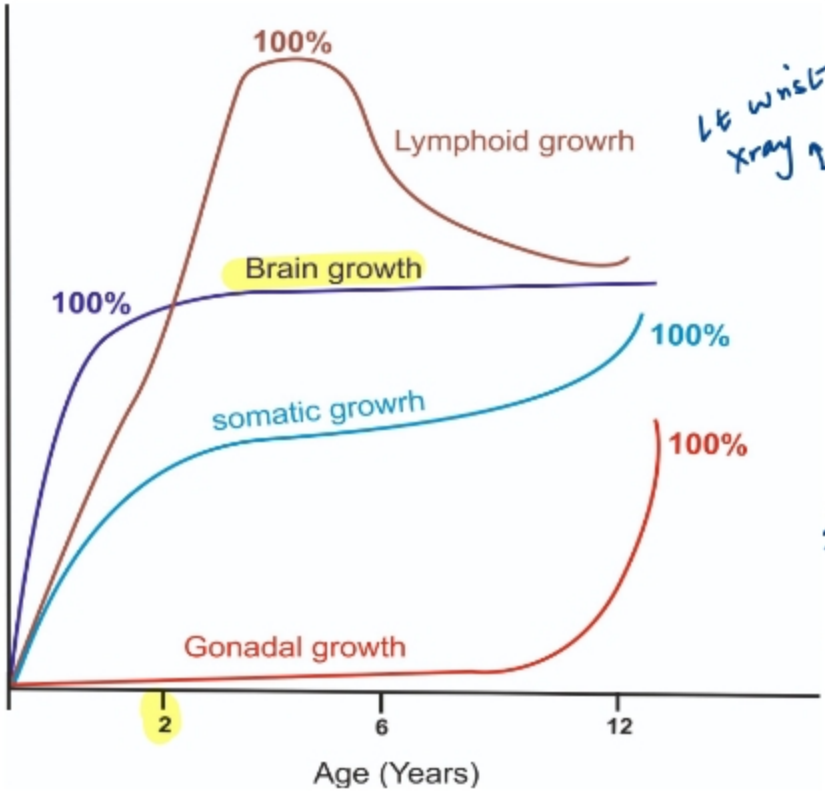
HC > CC by 2.5cm at birth
At 9-12mon: HC=CC → 47cm
>1yr: CC > HC

Surrogate marker of height: Arm span
- **Arm span < length by 2.5cm at birth**
- **Equal at 11yrs**
- **Arm span > length by 1cm after that**
- **Mid-parental height:** (Mother + father) / 2 ± 6.5cm (boys / girls)

Proportionate short stature: ↓ GH

Dispoportionate short stature-Short trunk — Δ facies; Hemi-vertebrae; Cong hepatic fibrosis
- **SED, MPS, Pott spine, Alagille Sx**

Dispoportionate short stature-Short limb
- **Rickets, Achondroplasia, OI, Congenital hypothyroidism**



SHORT STATURE: <-2SD / 3rd

Lt wrist xray

Bone Age < Chronological Age

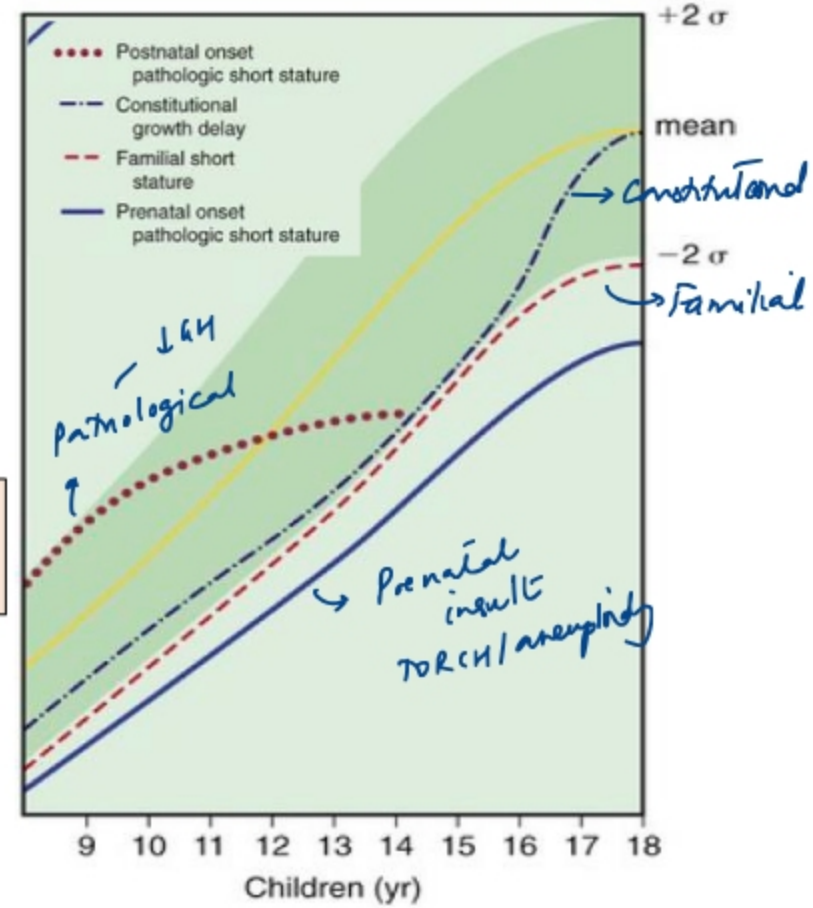
↳ Constitutional delay ♂♀ → Puberty delay

Bone = Chronological Age

Familial SS ↳ Puberty (N)

Hypogonadal GH ↓

Height velocity in CGD/Familial SS: (N)



Puberty: TPM

FEMALES:

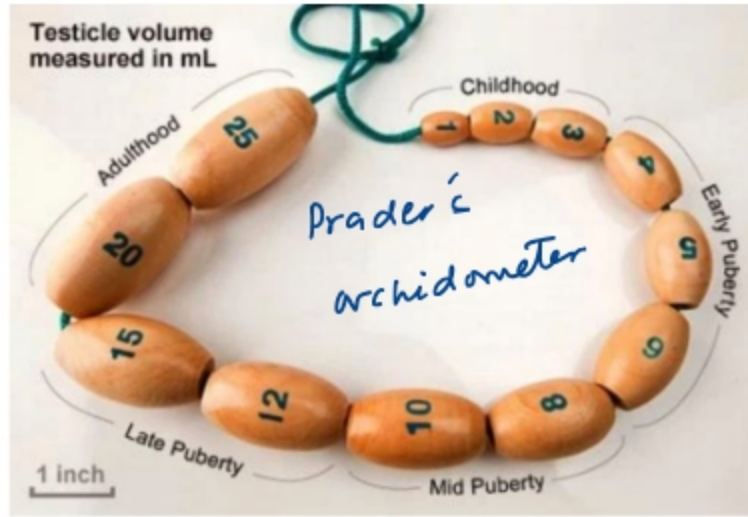
Thelarche
↓
Pubarche
↓ ← Growth Spurt (st 3)
Menarche

MALES:

Testicular enl
↓
Penile enl
↓ ← Growth Spurt (st 4)
Male voice / hair

Failure to thrive (FTT)
Descriptive term in <5yrs
Weight below the 3rd or 5th centile, failure to gain weight over time, or a change in the rate of growth such that weight for age or weight for length/height has crossed two major centiles (e.g., 50th to 10th) over a period of time

Tanner Stage	Male Genital Appearance	Female / Male Pubic Hair Appearance	Breast Appearance
Stage 1	Testicular volume < 3 mL	No pubic hair	Elevation of papilla only
Stage 2	Change in texture to scrotal skin	Sparse growth along the labia / base of penis	Breast bud stage
Stage 3	Increase in size of penis and testes	Darker, coarser, more curled hair	Enlargement of breast and areola
Stage 4	Further enlargement with development of glans penis	Adult-type hair over a smaller area	Projection of the areola and papilla
Stage 5	Adult size and shape	Spread to the medial surface of the thighs	Recession of the areola, projection of papilla only



Five Components of Nurturing Child Care

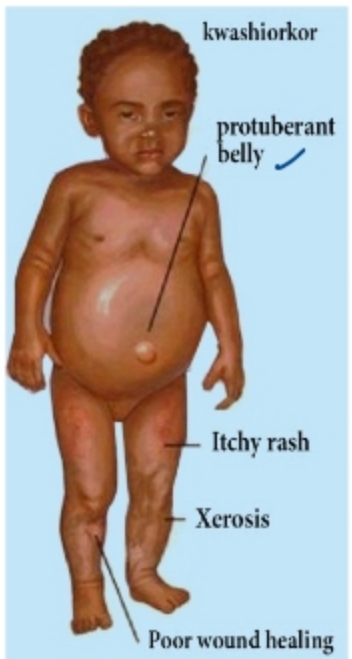
1. Good health
2. Adequate nutrition
3. Responsive caregiving
4. Security and safety
5. Opportunities for early learning

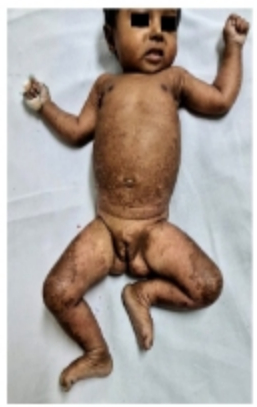
MALNUTRITION

Indicator	Parameter (<-2SD)	Interpretation
Stunting	Low height for age	Chronic malnutrn
Wasting	Low weight for height eg	Acute malnutrn
Under weight	Low weight for age	Acute on chronic

Severe acute malnutrition (SAM):
6-59 months of age

- Weight-for-height below – 3 SD of the median
- Visible severe wasting
- Presence of bipedal edema
- Mid-upper arm circumference below 11.5cm





Kwashiorkor	Marasmus
Deficient of protein Serum albumin: <3g/dl	Deficient of proteins and calories Serum albumin: >3g/dl
Subcutaneous fat preserved Triceps skin fold: >50th centile	Subcutaneous fat not preserved Muscle wasting Triceps skin fold: <3rd centile
Oedema Flaky paint dermatitis Flag sign Fatty liver	Oedema absent Loose, wrinkled skin Simian facies
Lethargic	Alert and irritable
Poor appetite	Voracious feeder

Management of SAM

Poor appetite / Edema ++ /Medical complications

HOSPITAL Mx-

S - Sugar - Hypoglycemia < 54mg/dl → iv dextrose bolus 2 ml/kg then infusion

H - Hypothermia

I - Infections

El - Electrolyte abN

De - dehydrated

D - Deficiency - vit / minerals - Fe - 2nd wk ✓

Dehydration in SAM best assessed by: urine output

Stabilisation: 0-7 days

F-75 (75 kcal + 0.9 g protein)/100 mL

All micronutrients except iron

Rehabilitation: 2-6weeks

F-100 (100 kCal + 2.9 g protein)/100 mL

Add iron

Primary failure: QP

- Failure to regain apetite by day 4
- Failure to lose edema by day 4
- Presence of edema on day 10
- Failure to gain at least 5g/kg/day by day 10

Breastfeeding

Constituent	Breast milk (gm/L)	Cow's milk (gm/L)
Proteins	11	33
• Casein	4	28
• Soluble proteins (whey)	7	5
• Taurine, cysteine	+	-
• DHA	+	-
Lactose	70	50
• Ca	0.33	1
• P (↑ soluble)	0.15	1
Vitamins		
• C	60mg	20mg
• D	501U	251U

Buffalo > Goat > Cow

Anti-infective:
TGF-B, Lactoferrin, IgA, Bifidus factor, Bile stimulated lipase
Colostrum: ≤3d – ↑↑ Ig → transitional → mature milk
Foremilk: water, protein, vitamins-mineral, carbs
Hindmilk: calories ← FAT
Adequate feeding:
Sleep- 2-3 hrs Urine- 6-8 times/d Gaining weight

Breast milk deficient in: vit D (400 IU at birth), vit K (2mg im), Fe ↓ ≥6mm
Max Breast milk output: 6mon
Expressed Breast milk: Room temperature 6hrs
Refrigerator 24hrs Deep freezer 3-6mon
Complementary feeding: Acceptable, Feasible, Affordable, Sustainable, Safe

Age (IYCF)	Quantity of feeds (n 250ml)	Frequency of feeds
6m-1yr	½ – 1 katori (a.a)	3 times /day
> 1 year	1 – 1½ katori	5 times /day

Adequate positioning:
Body well supported
Occiput, shoulder, buttocks in straight line
Entire baby turned towards mother
Abdomen touch-baby-mother

Adequate attachment:
Mouth wide open
Only small part upper areola visible
Lower lip everted
Chin touch mother's breast

Approach to rickets

Feature	Nutritional Rickets	Vit D–Dependent Type I (AR)	Vit D–Dependent Type II (AR)	X-linked Hypophosphat Rickets (XLD) (vit D Resist)	CKD
Defect	vit D ↓	1α OH ××	end organ R	FGF-23 ↑ (PHEX)	1α OH ⊖
Ca	↓	↓	↓	N	↓
PO_4	↓	↓	↓	↓ QQ	↑ QQ
PTH	↑	↑	↑	↑ N	↑
$1,25(OH)_2D_3$	↓	↓	↑↑	N	↓
Response to Vit D	✓✓	calcitriol ✓✓	↑↑ doses ∫ calcitriol	×× Rx – Burosumab	vit D ✓ Ca ✓ PO_4 binders

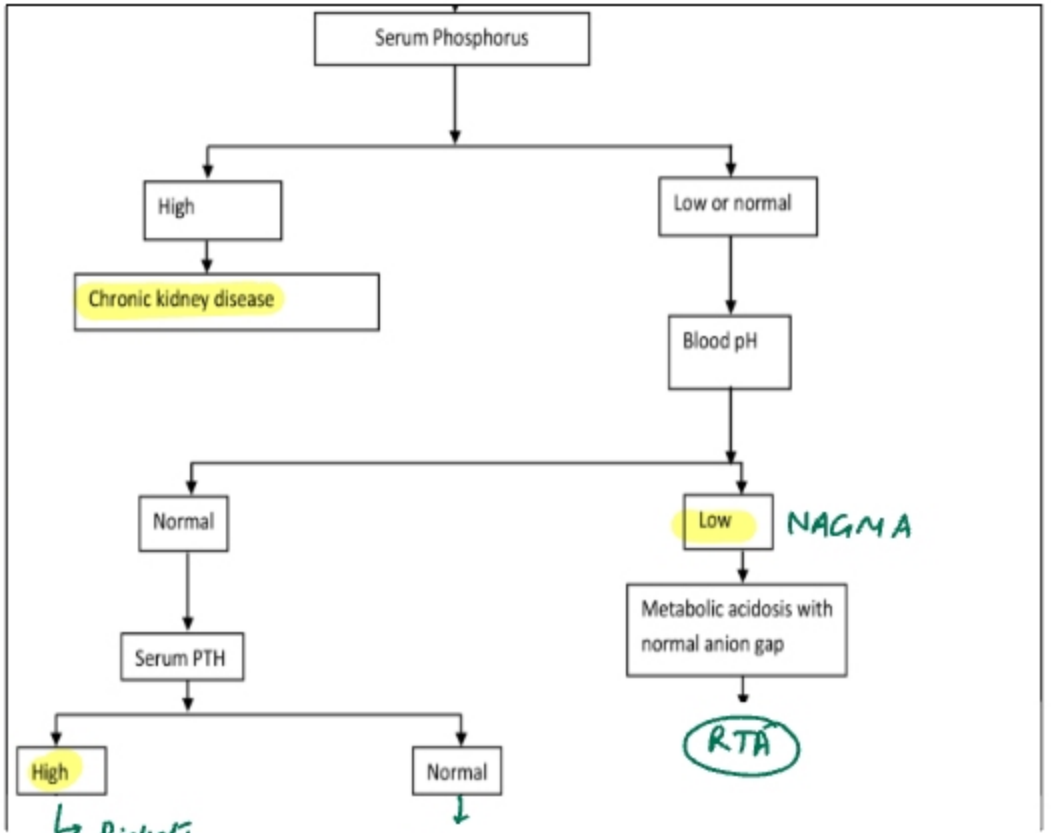
Rx - vit D
2000 IU x
12 wks

↓ FGF-23

Ca multiplying factor

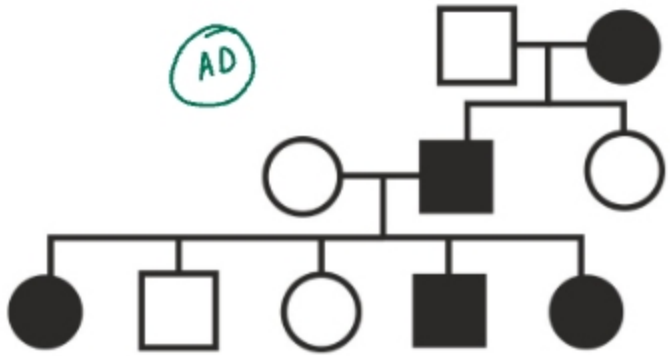
Ca × PO_4 >55
↓
(P) binders

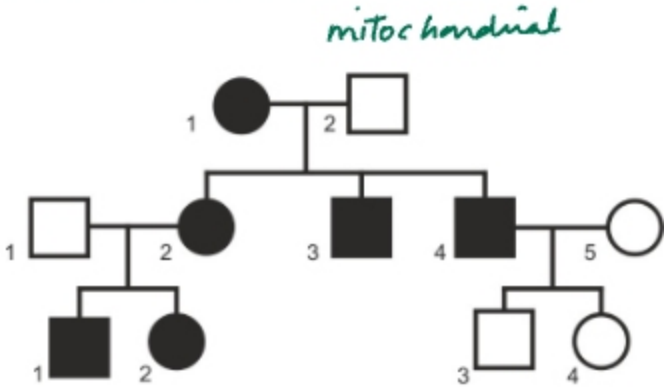
tumor induced osteomalacia → FGF-23 → Hemangiopericytoma / Hemangioma

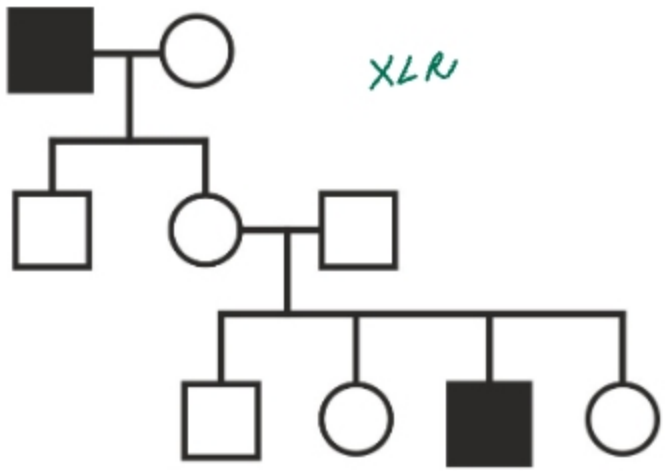


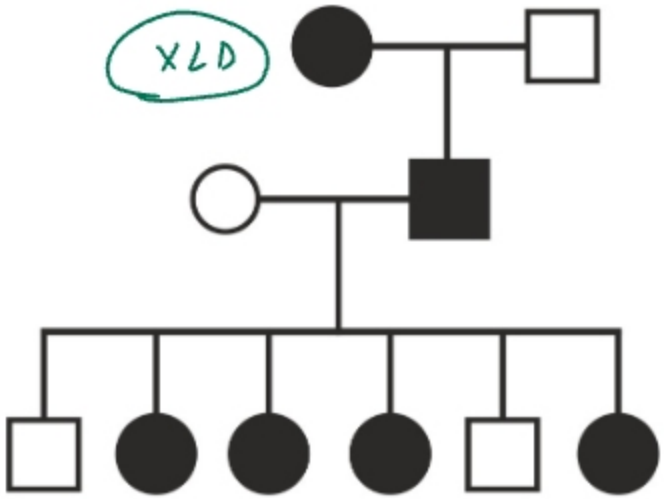
Pedigree Analysis

	STEPWISE:
AD (65%)	No gender pref All generations affected
AR (25%)	No gender pref Generations skipped
Mitochondrial	Mother to ALL - Heteroplasmy
Y-linked (Holandric)	Father to ALL sons
XLD	Father to all daughters Mother to 50% kids
XLR	Males affected, females carrier + Generations skipped
Gonadal mosaicism	Surprise case!









INHERITANCE

AD

HS, VWD (except type 3 / 2N)
Marfan, Achondroplasia/ EDS/ OI
AIP Noonan Sx
MEN, Neurocutaneous syndromes
Sturge Weber (sporadic) ATM (AR)

AR

All inborn errors of metabolism except Hunters / Fabry's (XLR)
Sickle cell anemia, Thalassemia
CF
Wilson, Hemochromatosis, Albinism, CAH

XLR ♂♀

G6PD
DMD/ BMD
Hemophilia A / B (C-AR)
Wiscott Aldrich/ Bruton's/ CGD
Lesch Nyhan
Lowe Sx
Colour blindness
Menke's ATP 7A
OTC deficiency
Barth Sx (Tafazzin)

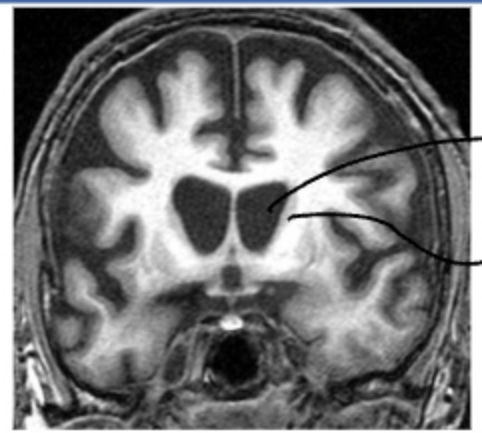
XLD

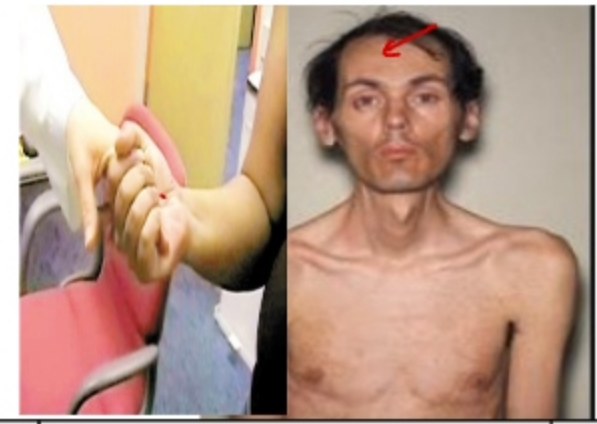
RP, Rett syndrome
Alport
Vit D resistant rickets
Incontinentia pigmentii

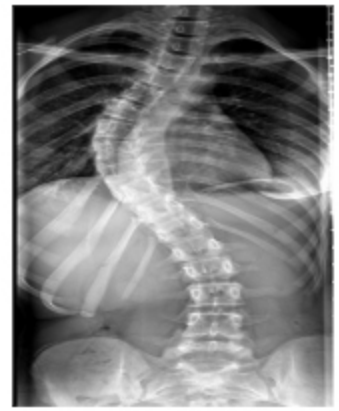
Mitochondrial (muscle, brain) — lactic acidosis

MELAS
MERRF ♡ cndn
Kearns-Sayre/ CPEO
NARP -neuropathy / ataxia / RP
LHON Leber's
Leigh Sx
Pearson Sx pancreatitis, sideroblastic anemia

Trinucleotide repeat disorders



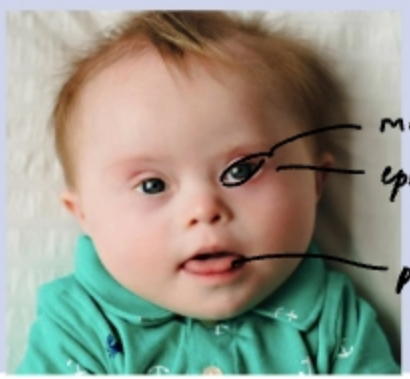
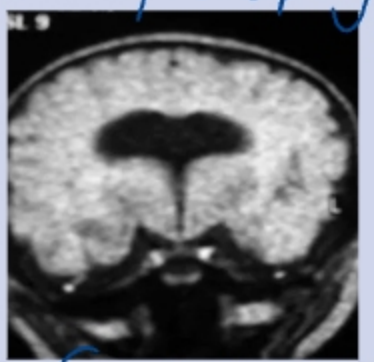
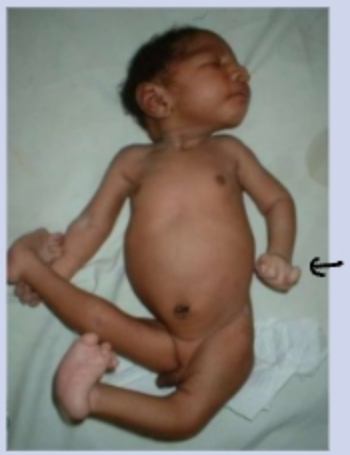




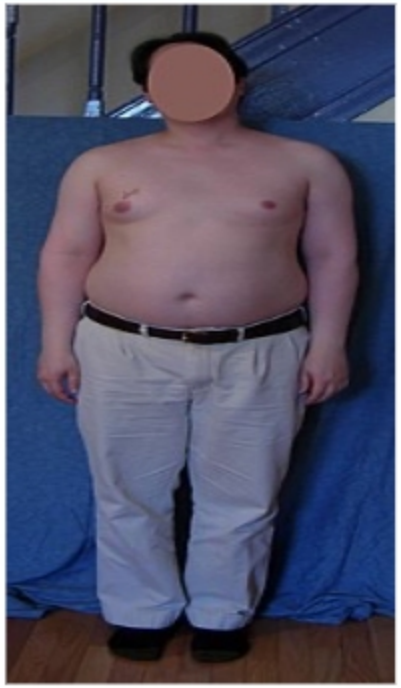
Diagnosis	Huntington chorea	Fragile X	Myotonic dystrophy	Friedrich ataxia
Repeat	C A G — coding ↓ caudate / / spinocer ataxia — kennedy Dopa ↑ Ach GABA	C G G (long giant gnads)	C T G (cannot terminate grip)	G A A
Inheritance	AD	XLR / XLD	AD	AR
Chromosome	4	X	19	9
Gene	Huntingtin	FMR1	DMPK	Frataxin
Others	- Sherman paradox } successive gen° earlier & severe - Anticipation	familial MR - mcc >200 repeats 55 - 200 premut° — ataxia, premature ovarian failure	- Hatchet facies - Christmas tree cataract	- ataxia ⊕ - DC ✓ - Neuropathy - spinocereb ✓ hallux valgus scoliosis

Rx - Omavexolone

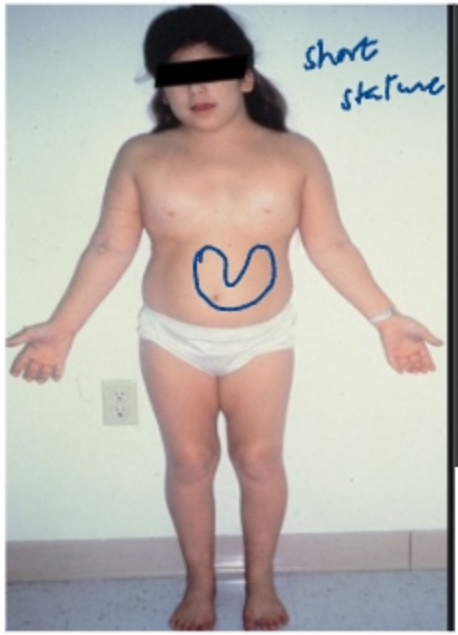
Genetic disorders- TRISOMY

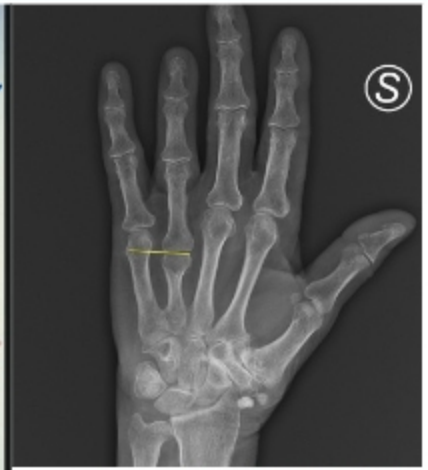
	DOWN (MC) 21	PATAU 13	EDWARD (2nd MC) 18
TONE	Hypotonia	Hypotonia	Hypertonia
HANDS	Simian crease / Clinodactyly	Polydactyly	clenched fist ± overlapping
FEET	Sandal gap	Rocket bottom feet	———
EYES	Brushfield spots - iris	Hypotelorism (cyclops) / PHPV	ophthalmia
CVS	Endocardial cushion - AVSD	VSD	VSD
MOUTH	Protruding tongue	CL/CP	CL/CP
OTHERS	MC genetic cause of Low IQ GI: Duodenal atresia > HD AAD, Hypothyroid, Alzheimer's, ALL, AML-M7 (≤3yr) CHL - serious om Basilar invagin / Grisel Sx Maternal Meiotic non-disjunction (95%) Robertsonian translocation-3% - t(21;21) (100% chance of recurrence)  mongoloid slant epicanthal folds protruding tongue mid facial flattening	Not with maternal age PHPV Aplasia cutis  Holoprosencephaly  (pancake brain)	Low IQ Maternal age Abdominal wall defects Horseshoe kidney 

SYNDROMES

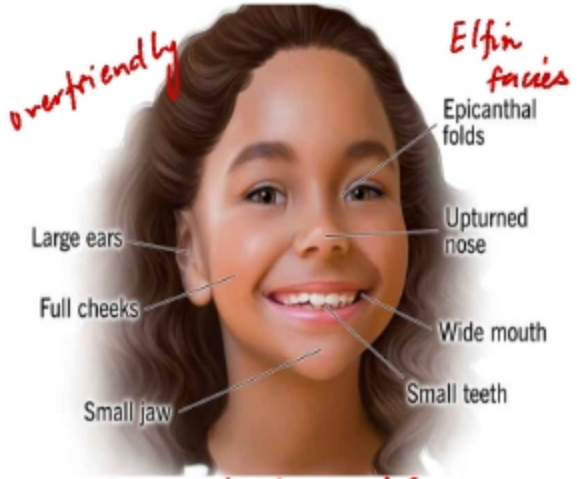


tall ht





turner's → 4th mc



- supravalvular AS
- ↑Ca2+

William Sx

"chrill by"

cri-du-chat

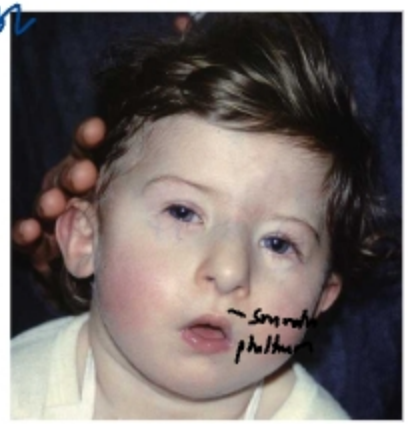
Turner's (XO) ↑rlo

Mosaic: XO; XY → gonadectomy gonadoblastoma
Lymphedema of feet and hands
Webbing of neck, Cystic hygroma
Shield chest, Inc carrying angle
Gonads: streak ovary
CVS: Bicuspid aortic valve > CoA
SNHL
IQ: (N)
NOONAN(AD): (N) XX/XY + phenotype turner's
IQ (↓) - MR
Pulm stenosis

Klinefelter (XXY)

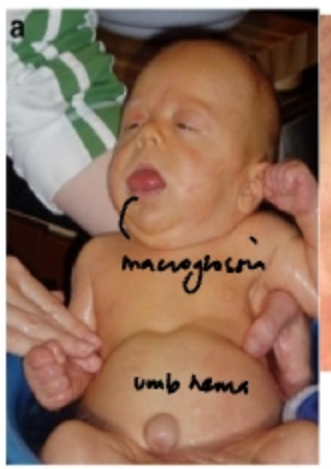
Non-disjunction of X
Paternal age
IQ: MR ≅
Gonads: atrophy
Gynecomastia
Sparse hair

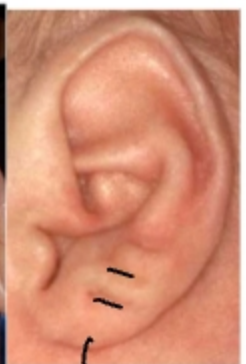
(↑gonadotrophic hypogonad)
↑FSH ↑LH
Testost ↓



DiGeorge Sx - 22 del^{n}

↓ Ca2+ — T cell dysfn.





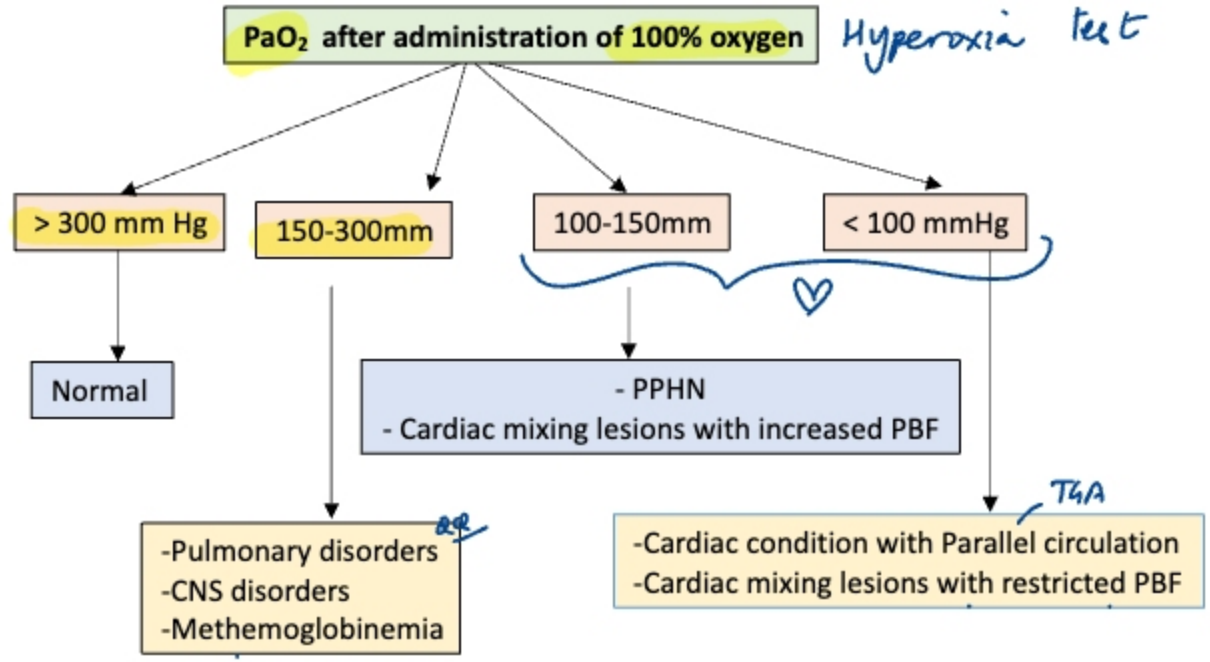
IUGR — WT

Beckwith Wiedemann — 11

Congenital Heart Diseases-Approach

-MC overall: VSD
-MC to be affected by IE: VSD
-MC cyanotic HD: TOF
-MC cyanotic HD in neonates: TGA / TAPVL - obstructed / Infracardiac
-MC cause of death in first week: HLHS

NADA'S CRITERIA	
MAJOR	**MINOR**
1. Systolic murmur Grade ≥ 3 2. Diastolic murmur 3. Cyanosis 4. Congestive Heart Failure	1. Systolic murmur ≤ Grade 2 2. Abnormal Second hear sound 3. Abnormal ECG 4. Abnormal Chest Xray 5. Abnormal Blood pressure



Acyanotic CHD

L → R

R → L shunt "Eisenmenger Ex"
Reversal

PULMONARY PLETHORA – Pbf ↑↑

2nd heart sound wide and fixed split

ASD

ins A2 – P2
Exp A2 – P2

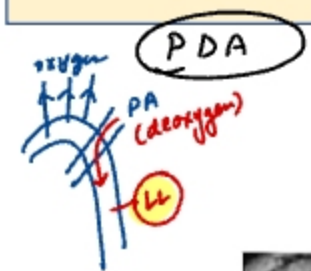
- **RAD** → ostium secundum (mc)
- **LAD** → primum

Pansystolic murmur (tricuspid area)
LAD
MC type: membranous

VSD

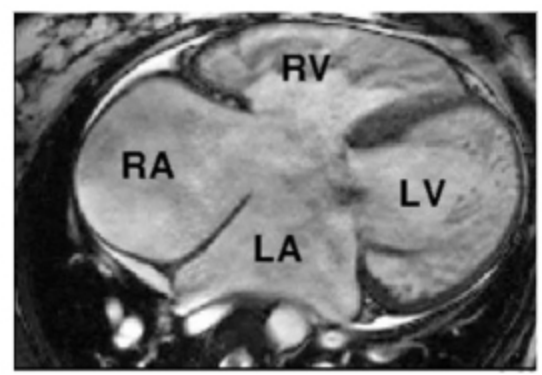
mc cong HD

Continuous, machinery infraclavicular murmur
Differential RR cyanosis → PDA + Eisenmenger



Goose neck deformity
MC in downs

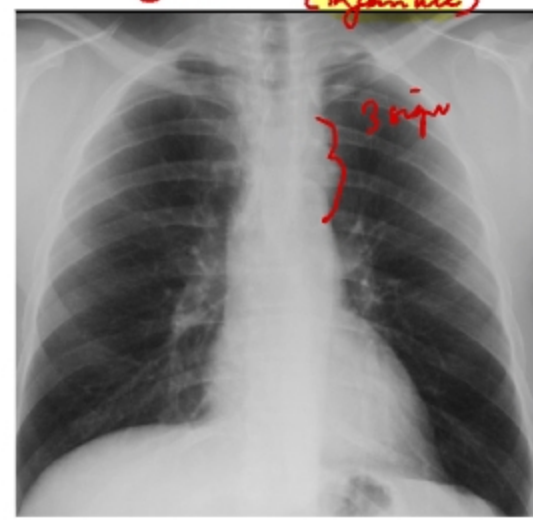
AVSD / Endocardial cushion defect

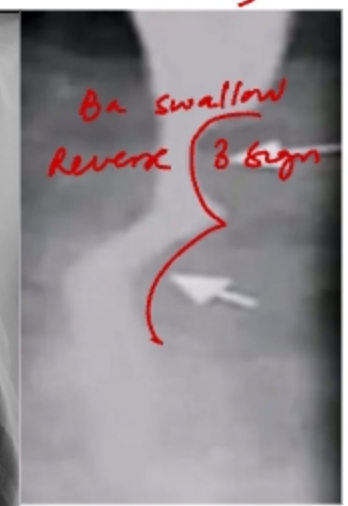


Normal PBF

Brachio-femoral delay
Intermittent claudication
Hypertension

Coarctation of Aorta
(x) collaterals
Pre-ductal (infantile)
Post-ductal (mc)





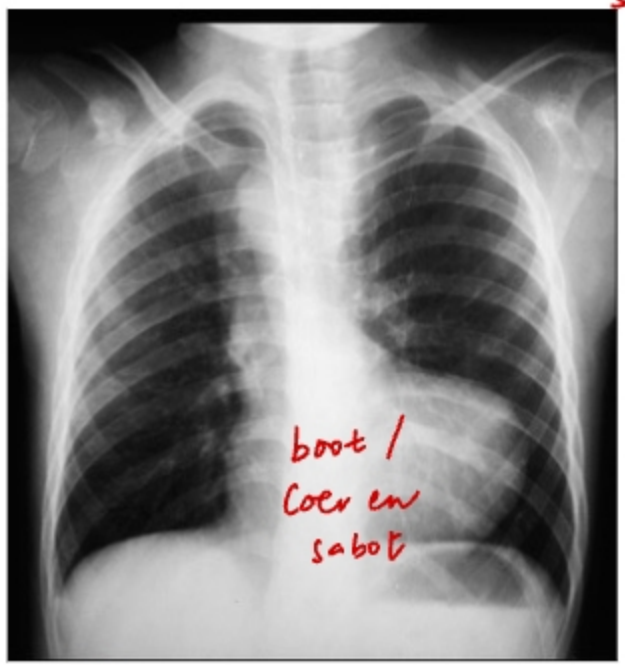
Int mammary → ant ic → post ic → desc Ao
2 SCA
3–9th ic

inf rib notching = Roesler sign

Cyanotic + Oligemia

R → L shunt (Pbf ↓)

RAD (RVH) – Pulm stenosis
Pulmonary ejection systolic flow murmur
Single S2 (A2 =)

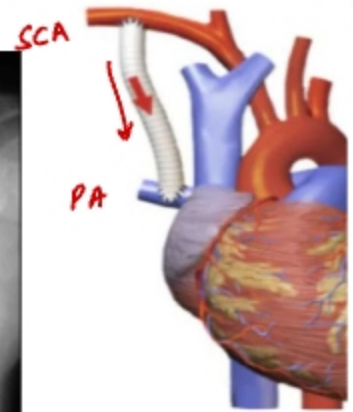


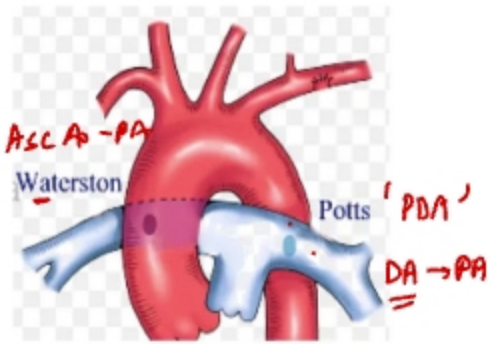
TOF
- RVH – upturned apex
- Pulm stenosis – oligemia
- VSD (R → L shunt) – ⊗ ♡ failure
- overriding of aorta

Pentalogy: + ASD

PA
Aorta

Blalock – Taussig



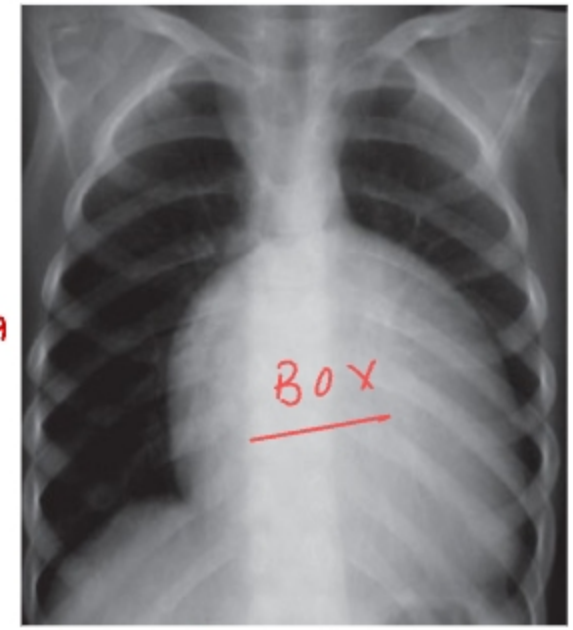


Cyanotic spells Mx: ↓↓ Pbf

1. knee-chest / squatting ↑ afterload / SVR
2. Morphine
3. O_2
4. β-blockers – ↓ spasm – ↑ Pbf

xx v

WPW, RBBB, Himalayan P waves



"atrialisation of RV"
oligemia

Ebstein anomaly

Li

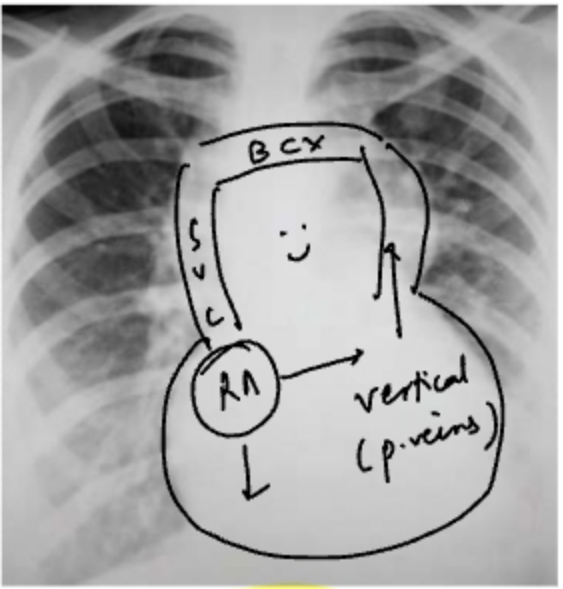
LAD

Tricuspid atresia

Cyanotic + Plethora

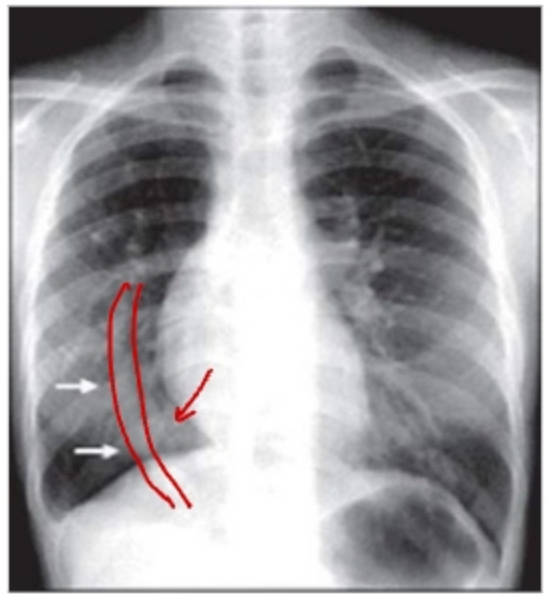
Pbf ↑

LARGE PEDICLE
2ND HS WIDE AND FIXED
Same saturation in all chambers



TAPVC

- Supracardiac (I) — MC
  - figure of 8 / cottage loaf / snowman
- Cardiac
- Infra-Cardiac
  - worst prognosis
  - obstruction → p. edema ++ cyanosis



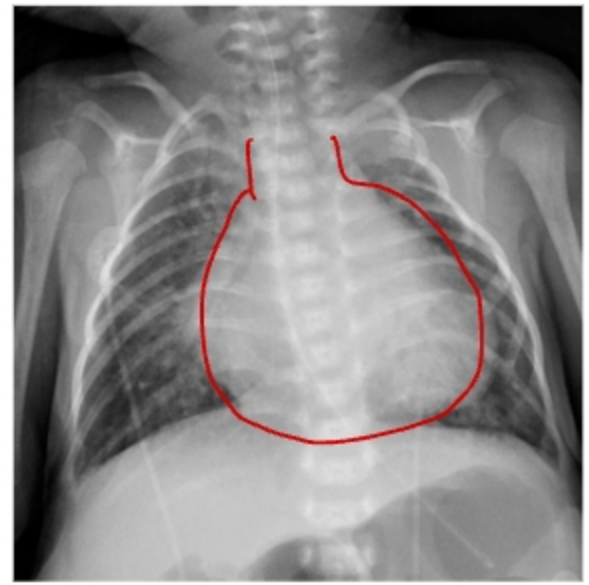
PAPVC

(Rt p.veins → IVC)

Scimitar sign

Rt lung hypoplasia

SMALL PEDICLE



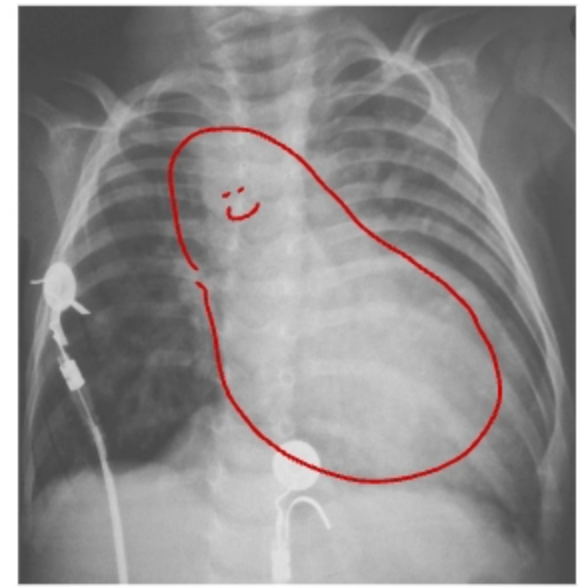
"egg on string"

TGA

- septum-dependent
  - ↓
  - Alprostadil (PGE_1) → PDA ≡
  - Atrial septostomy ↓

Jatine's arterial switch

LARGE PEDICLE



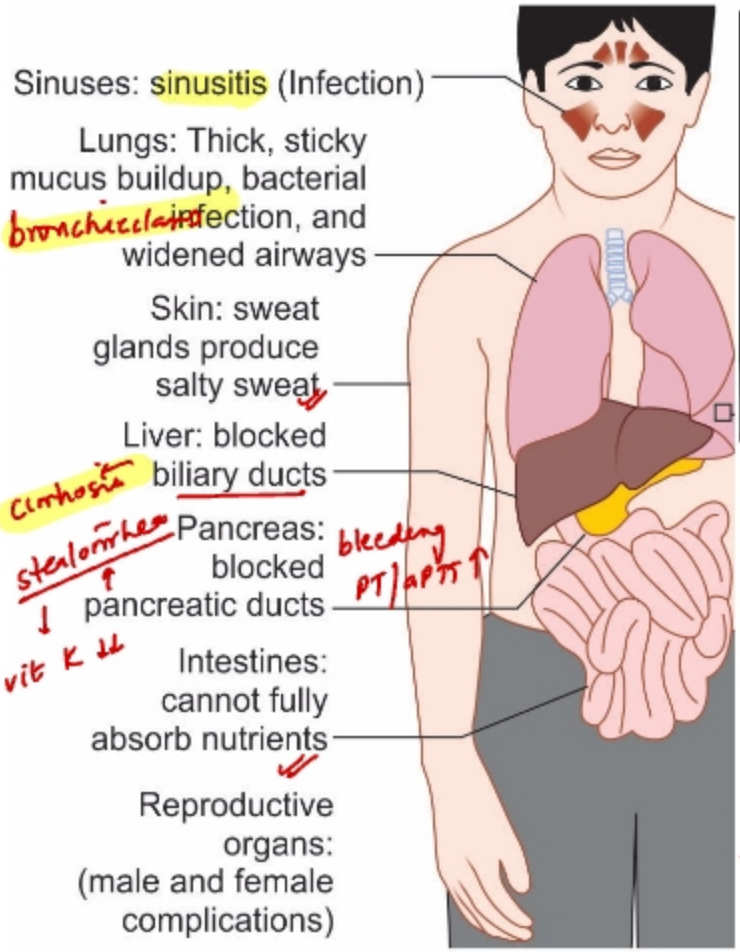
Sitting duck

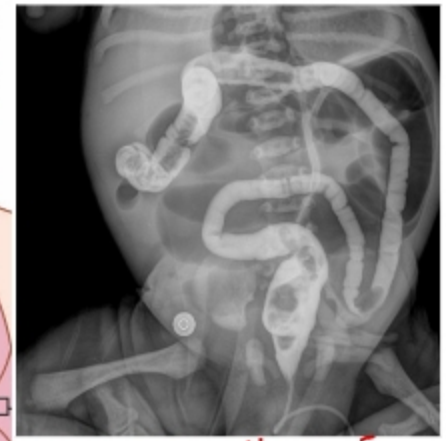
↓

PTA

Persistent truncus arteriosus

Cystic fibrosis





meconium ileus ± microcolon

dx + tx – Gastrografin enema

Bishop–Koop sx

Cystic Fibrosis Finding	Biochemical Evidence of CFTR Dysfunction
• ≥1 Phenotypical finding • Positive neonatal screening: Immunoreactive trypsinogen • Positive family history	• Positive sweat chloride >60meq/l on 2 separate days • Positive Nasal potential difference • 2 mutations in CFTR

CF: Chromosome: 6 – AR CFTR – Cl^- – misfolded prtn
-MC mutation: ΔF508
-MC class of mutation: class 2 - trafficking
-Trikafta:
Elexacaftor + Tezacaftor + Ivacaftor
(Elexacaftor + Tezacaftor: correction; Ivacaftor: potentiator)

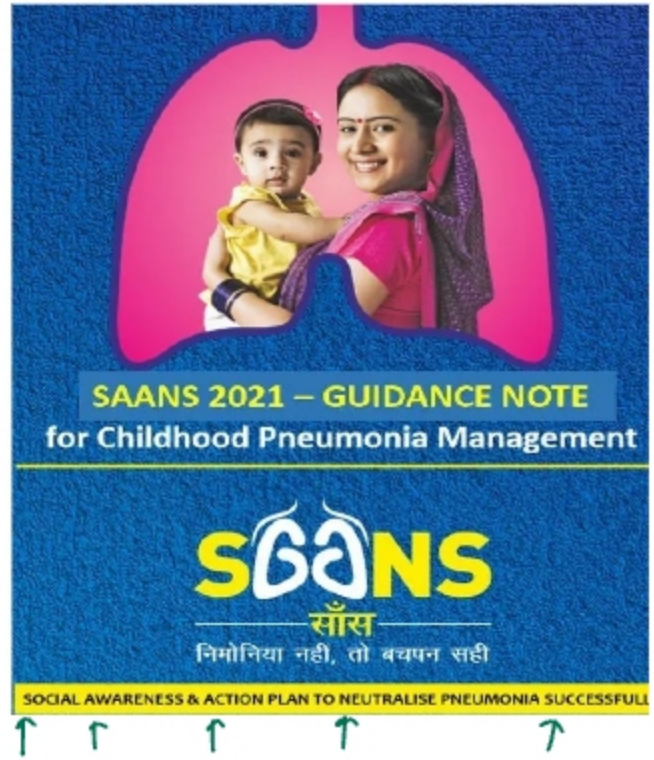
0-12yrs: Malabsorption/ steatorrhea MC
>13yrs: Pneumonia MC
Azoospemia in 98%

<16yr – S.aureus
>16yrs – Pseudomonas ← inhaled tobramycin
most sp – Burkholderia

CF → sweat glands → Cl^- levels: ↑
other ducts → ↓ : obstruction
Cl^- transport: ↓

Pneumonia

IMNCI Category	Features	Management
No pneumonia	Fever, cough/cold	• Symptomatic • Follow-up after **5 days**
Pneumonia	• Fast breathing (0-2m - ≥60, 2-12m - ≥50, >12m - ≥40) +/- chest indrawing	• **Oral amoxicillin × 5 days** • Follow-up after **2 days**
Severe pneumonia	• SpO_2 < 90% • Stridor in calm child • ≥1 danger sign: Lethargy, Not feeding, Convulsions, Cyanosis, unconscious, head nodding	• **Urgent referral** • Give **1st dose ampicillin + gentamicin im injection**



Acute Diarrhea

Parameters	No Dehydration	Some Dehydration	Severe Dehydration
Appearance	Well, alert	Restless, irritable	Lethargic, unconscious
Eyes	Normal	Sunken	Very sunken
Thirst	Drinks normally, not thirsty	Thirsty, drinks eagerly	Drinks poorly or not able to drink
Skin pinch	Goes back quickly (<1 second)	Goes back slowly (1 second)	Goes back very slowly (2 seconds)

ZINC: 14days
<6mon: 10mg
>6mon: 20mg

↓
fluids orally
ORS

75ml/kg over 4 hours
↓
ORS

RL + 5% dextrose
100ml/kg
30ml/kg / \ 70ml/kg

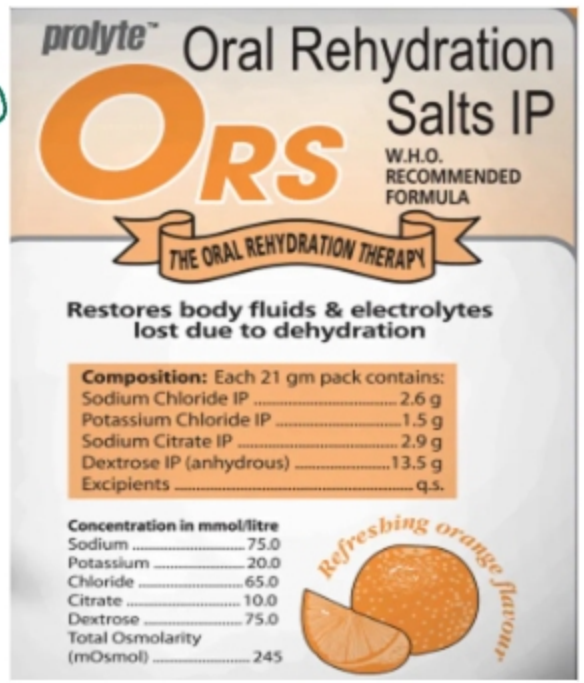
<1yr : 1hr 5hrs
>1yr : 1/2 hr 2 1/2 hrs

FLUID COMPOSITION

NAGMA = ↑Cl⁻ acidosis ∴ preferred in vomiting
∴ preferred in RL
xx in cirrhosis

Composition	ReSoMal (mmol/L) SAM	Standard ORS (mmol/L)	Reduced osmolarity ORS
Glucose	125	111	75
Sodium	45	90	75
Potassium	40	20	20
Chloride	70	80	65
Citrate	7	10	10
Magnesium	3	---	---
Zinc	0.3	---	---
Copper	0.045	---	---
Osmolarity (mOsm/L)	300	311	245

	Plasma*	0.9% NaCl NS	Hartmann's RL
Na^+ (mmol/l)	135–145	154	131
Cl^- (mmol/l)	95–105	154	111
$[Na^+]:[Cl^-]$ ratio	1.28–1.45:1	1:1	1.18:1
K^+ (mmol/l)	3.5–5.3	0	5
HCO_3^- / Bicarbonate precursor (mmol/l)	24–32	0	29 (lactate) ↓ Liver HCO_3^- (∴ metab alkalosis)
Ca^{2+} (mmol/l)	2.2–2.6	0	2
Mg^{2+} (mmol/l)	0.8–1.2	0	0
Glucose (mmol/l)	3.5–5.5	0	0
pH	7.35–7.45	4.5–7.0	5.0–7.0
Osmolarity (mOsm/l)	275–295	308	278



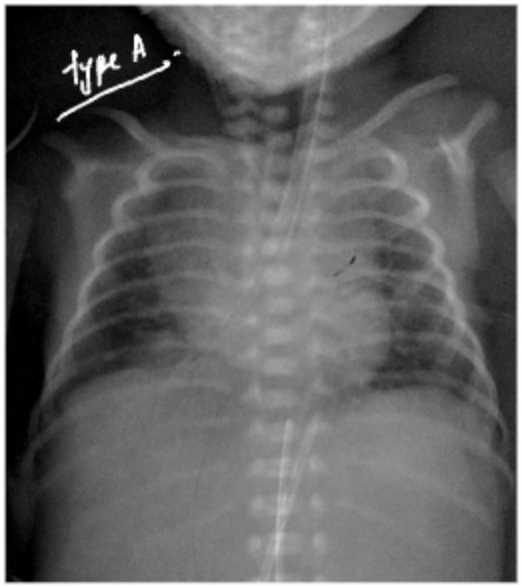
Fluid for maintenance: Holliday - Segar formula
N + 5% dextrose aa

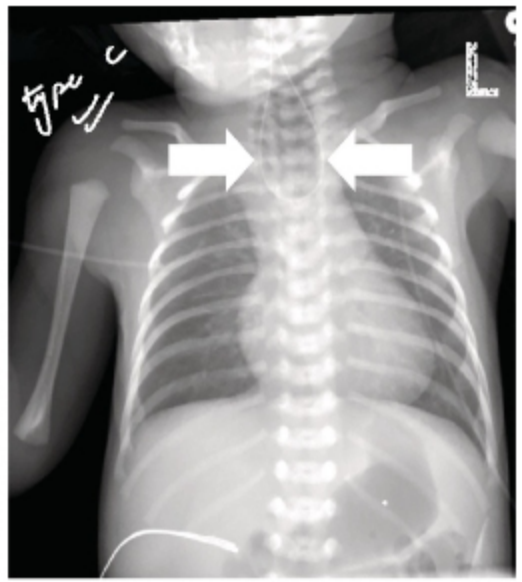
30 kg → 1000 + 500 + 200 = 1700 ml

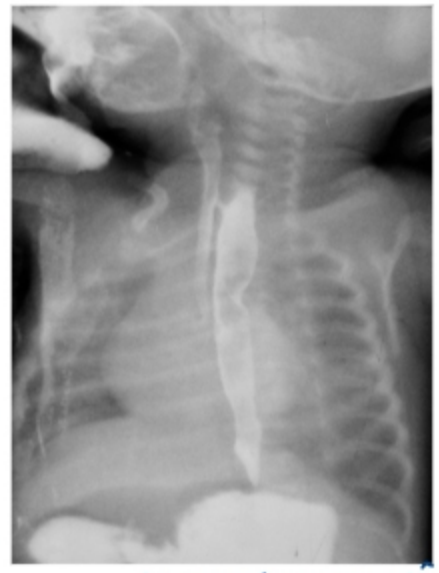
1st 10kg: 100ml/kg **Next 10 kg: 50ml/kg** **>20kg: 20ml/kg**

GI emergencies

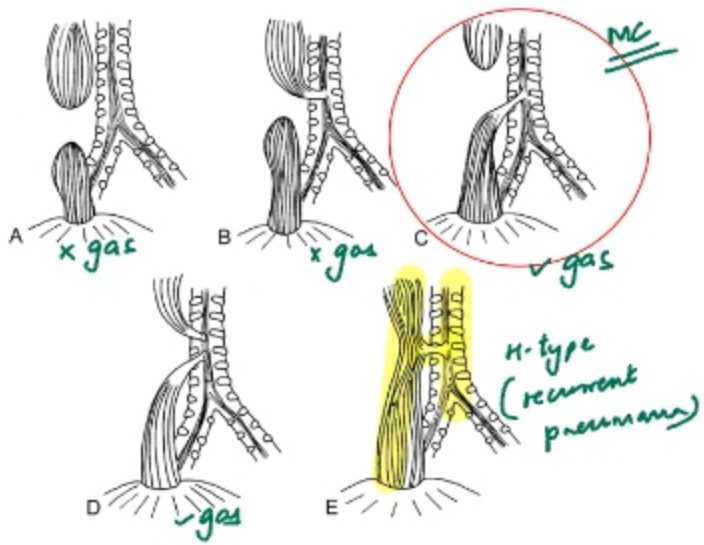
Newborn with drooling and difficulty feeding (TEF)




type C

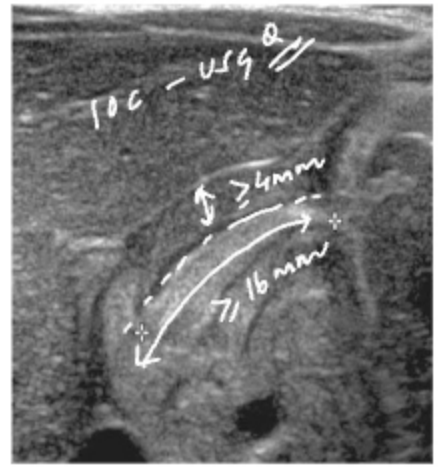


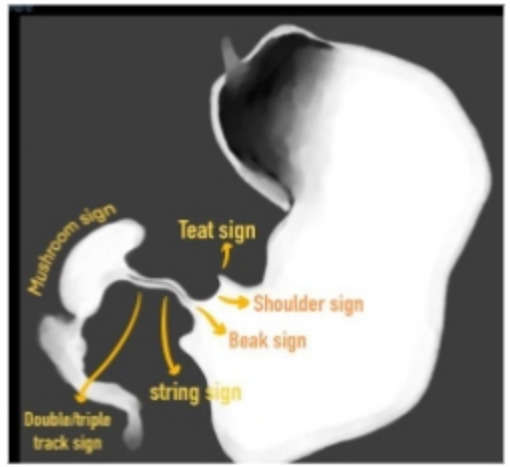
IOC → Gastrografin study.



- ass ε VACTERL
  - vertebral
  - Anal
  - Cardiac
  - TEF
  - renal
  - Limb (radial ray)

Non-bilious vomiting
Non-projectile- GERD
Sandifer Sx – posturing – arch back
Projectile- CHPS

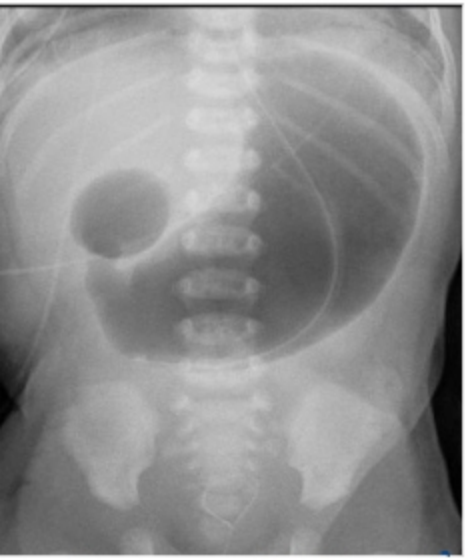




- olive shaped mass at LUQ
- L → R peristalsis
- 4-6wks after birth
- Maternal macrolides ++

Rx: Metab alkalosis ↓Cl ↓K+ ε paradoxical aciduria
↳ RAAS ⊕
correct.
↓
Ramstead pyloromyotomy / POEM

Billious vomiting

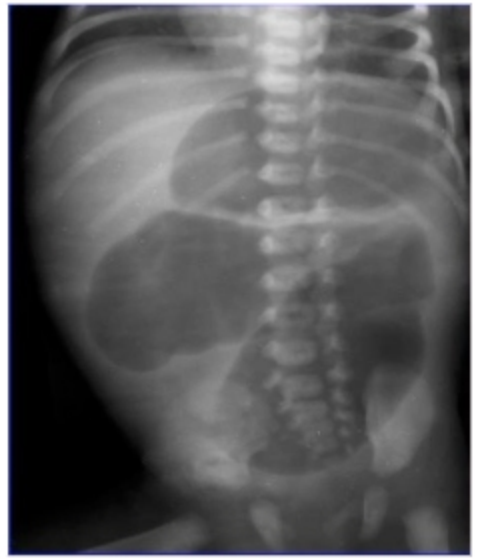


Double bubble sign

↓

- Duodenal atresia (Down's)

D/D
- Annular pancreas
- Ladd bands

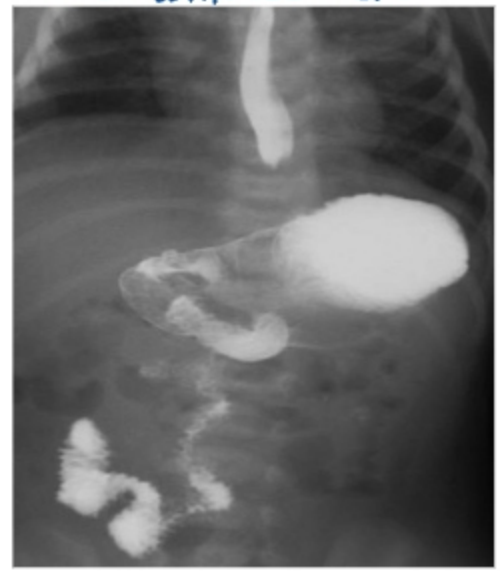


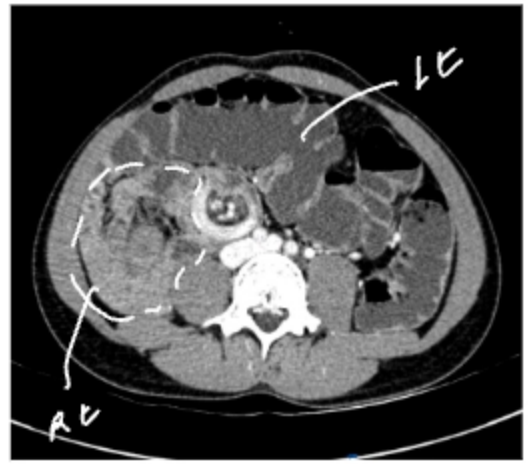
Triple bubble

↓

Jejunal atresia

UGI contrast stn



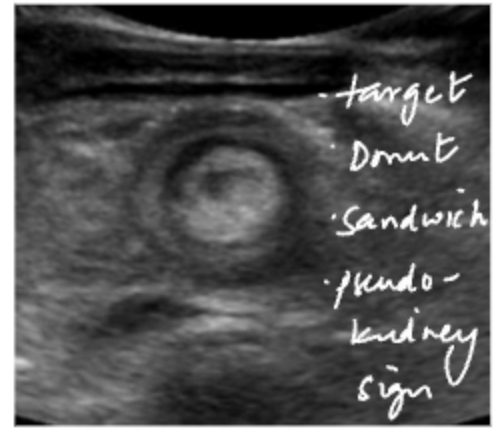


Corkscrew sign /whirlpool sign

Midgut volvulus ± malrotation

Pain + red currant jelly stools Intussusception

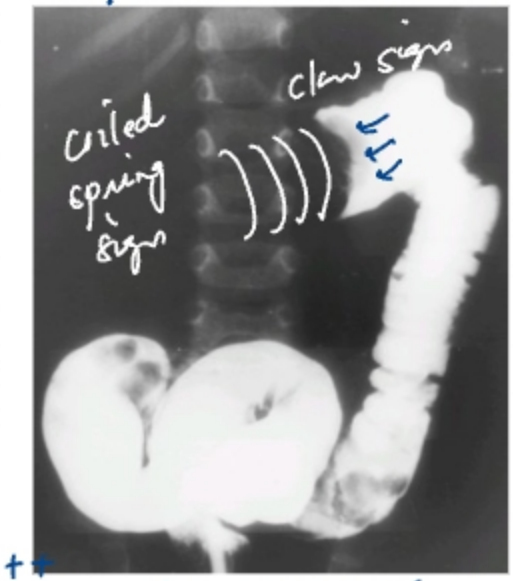
1st – USG



- 6m - 1yr
- Rotavirus / weaning ++
  ↓
  peyer's patches hypertrophy
- Ileo-colic (mc)

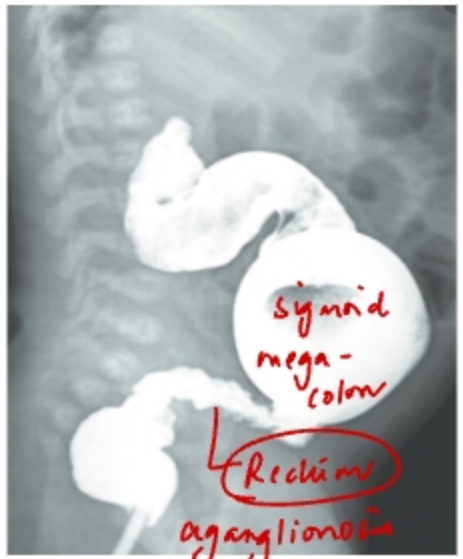
Adult → 'lead point'
↳ polyp / carcinoma / Meckel's / HSP

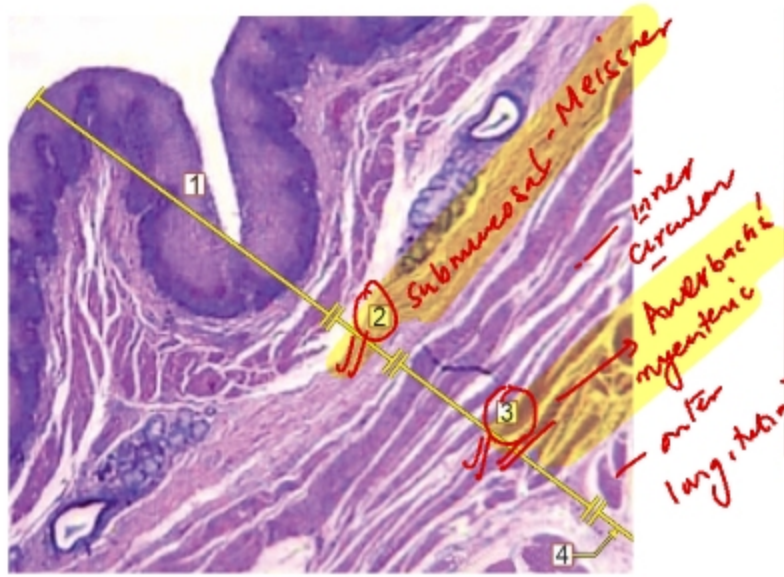
IOC / Gold std - dx + Rx



Ba enema / air / NS enema

Not passed meconium >48hrs after birth





Hirschsprung's D

→ Neural crest cells migrn ↓↓ → Nerve plexus ↓↓ → peristalsis ↓↓

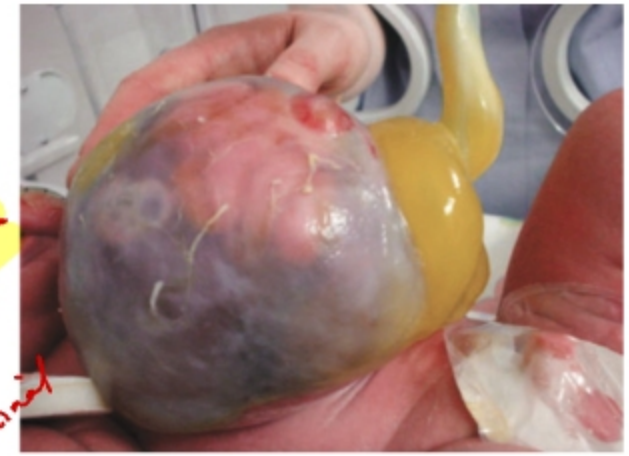
Rectum ↓↓

Ict → Ba enema → transition zone
└ Rectum - sigmoid ratio <1

IOC → Punch bx - AchE

Rx - Pull through procedure — Swenson / Soave / Duhamel

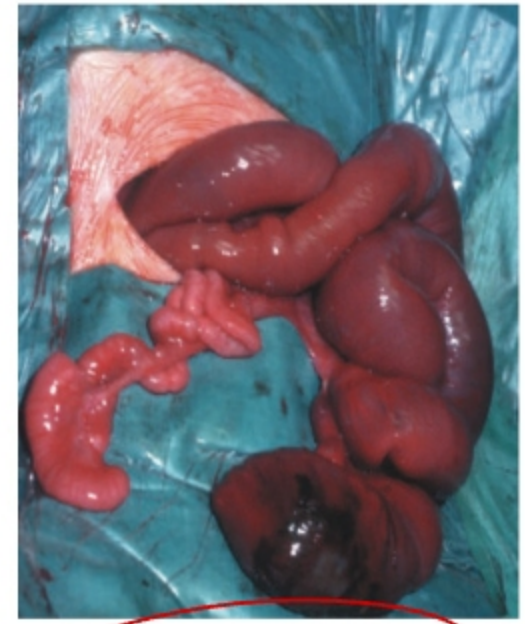
At birth



Peritoneum
Amnion
Wharton's jelly
Minor:<5cm

Omphalocele

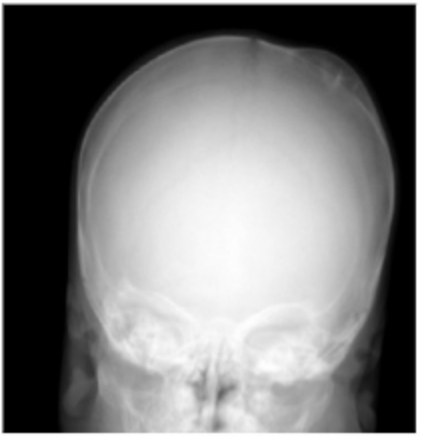
- membrane
- apex of umbilicus
- ↑ assn - cong anomalies

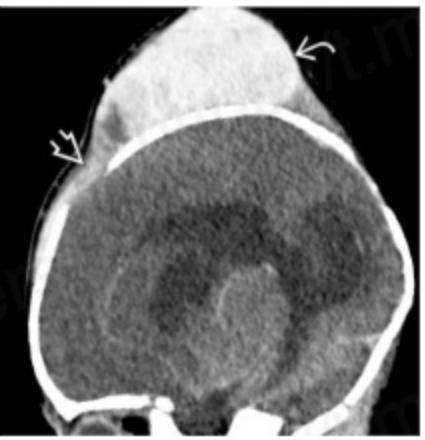


Gastroschisis

- off - midline
- ↑ necrosis - bowel

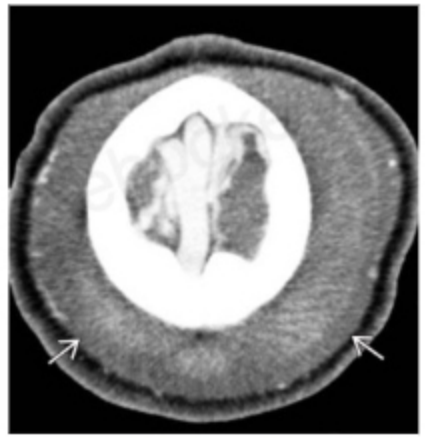
Skull pathologies



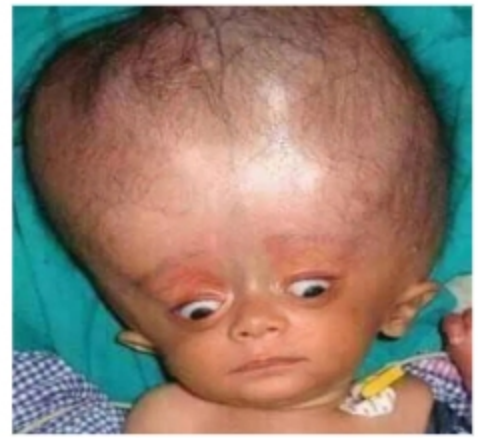


- Focal - subperiosteal
- Cephal hematoma - may ↑ in size
- prolonged jaundice

Caput succedenum - focal edema
- max size at birth
- Reassure

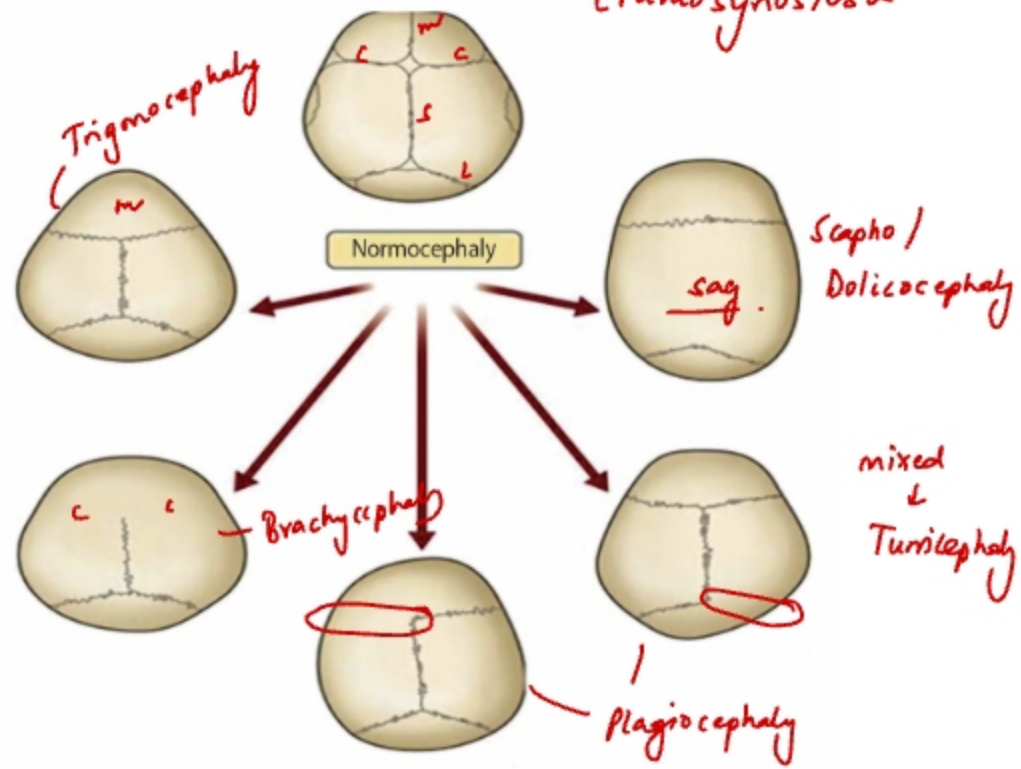


Subgaleal hematoma
↓
↑↑ bleed - Hypovolemic shock
- Diffuse



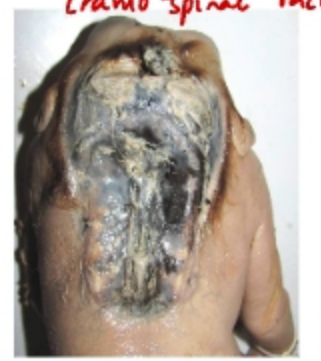
MCC: Aqueductal stenosis
Sunset sign – tectal plate compr
McEwen sign – crack pot
Mx: VP shunt
IOC for shunt infection:
Shunt tap

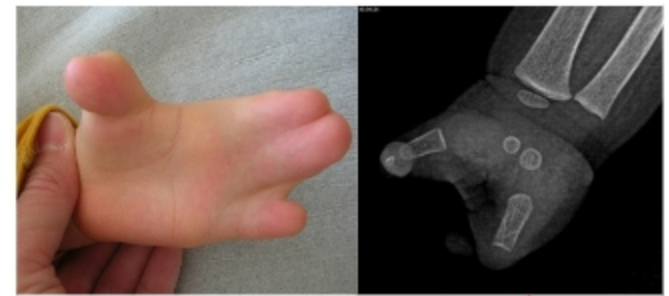
Craniosynostosis



Iniencephaly

Cranio spinal rachischisis



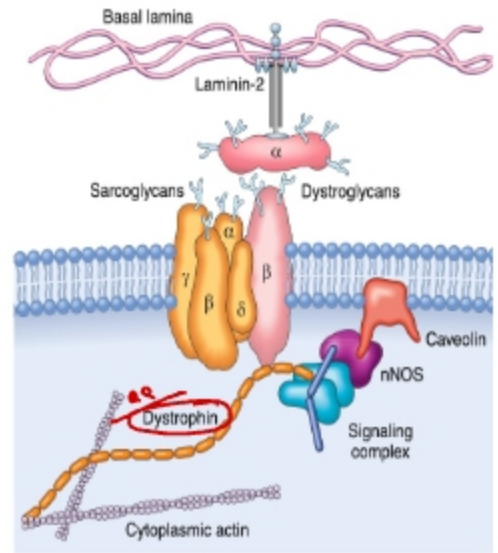


Apert Sx - syndactyly = Mitten hands
Crouzon Sx - mid face hypoplasia - FGFR2
Carpenter Sx
Pfeiffer Sx

Miscellaneous

Febrile seizures: Fever (>100.4) + seizure in 6mon-6yrs
- **Simple-GTCS, <15min, No recurrence in 24hrs**
- **Complex** - poor prognosis

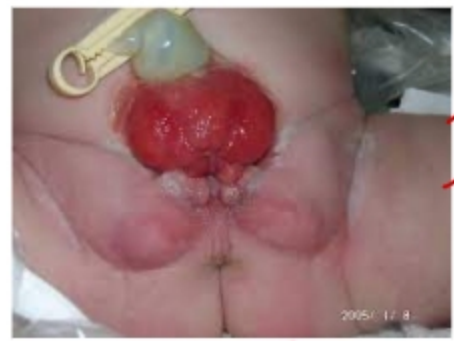
No long-term AED
If >5min: BZD DOC - Lorazepam / Rectal diazepam / inasal Midazolam
High risk for recurrence – 30-50%
Risk factors:
MAJOR: <1yr, <24hr fever, 100-102 F
MINOR: Complex, Family history , Low Na, Male, Daycare



Schwartz

$$\text{eGFR} = \frac{\text{Length (cm)} \times k\,(0.41)}{\text{Serum creatinine (mg/dL)}}$$

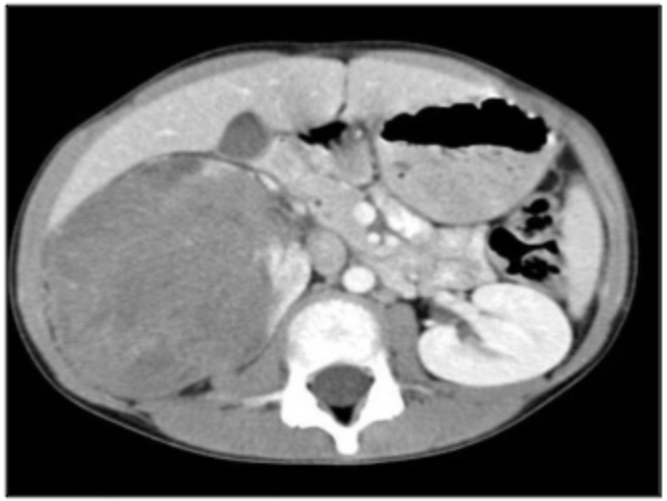
Congenital hypothyroidism
Growth retardation, umbilical hernia, Large tongue, Epiphyses delayed QQ
MCC: dysgenesis > dyshormogenesis
Screening: TSH → ≥48hrs of birth - DBS / heel prick
Next: USG thyroid / RAIU scan QQ



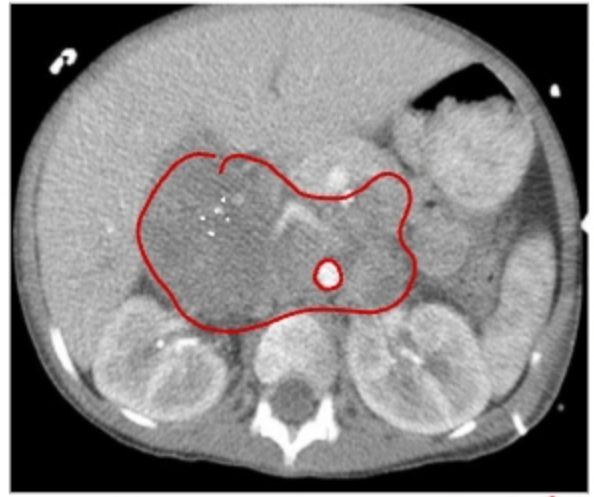
exstrophy - Ectopia vesicae - UB adenoca
Epispadias

late - ll
CPK very high, Dystrophin gene
Frameshift / Non-sense: absent dystrophin - DMD
In-frame mutation: ↓↓ dystrophin - BMD Milder
Cardiomyopathy, Low IQ
MCC of death: Resp muscle weakness
Eteplirsen - exon 51
Casimersen - exon 45

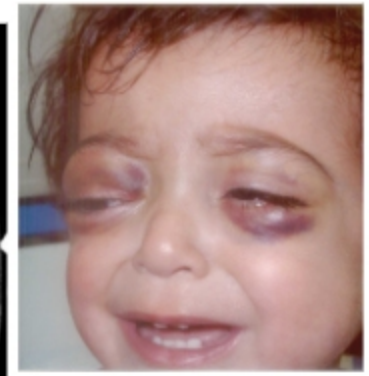
Wilms tumor VS Neuroblastoma



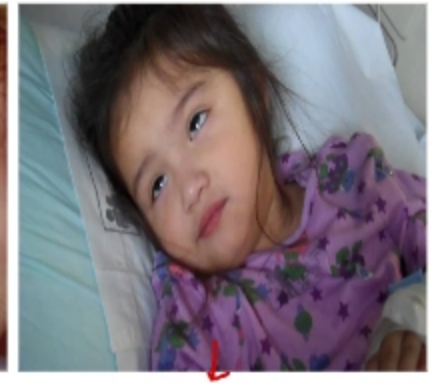
- 0-5yo · Flank mass
- invasion - RV / IVC
- mets - mc Lungs



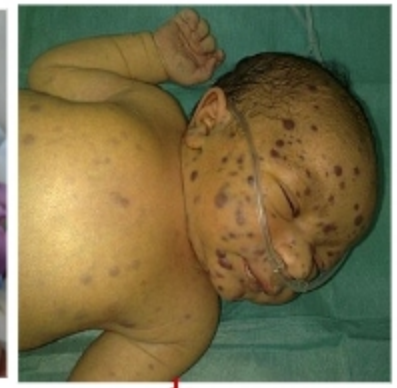
- RP → midline · Ca^{2+} ↑↑
- encase vessels
- mets - mc → Bones



Racoon eyes ↓ mets to orbit



opsoclonus - Myoclonus



Skin - Blueberry muffin

Risk factors:

WT: Chr 11

11p13: WAGR → WT / aniridia / Growth MR / GU anomalies

Denys Drash → mesangial sclerosis / GU

11p15: Beckwith Weidmann

Horseshoe kidney

UDT, Hypospadias

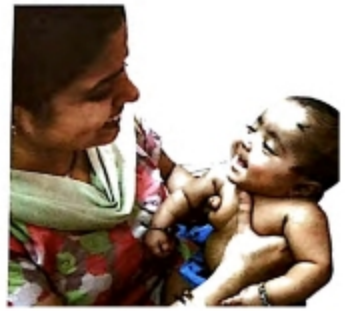
NB

- **Hypodiploidy**
- **N-myc amplification**
- **ALK amplification**
- **Loss of heterozygosity-1p,11q**

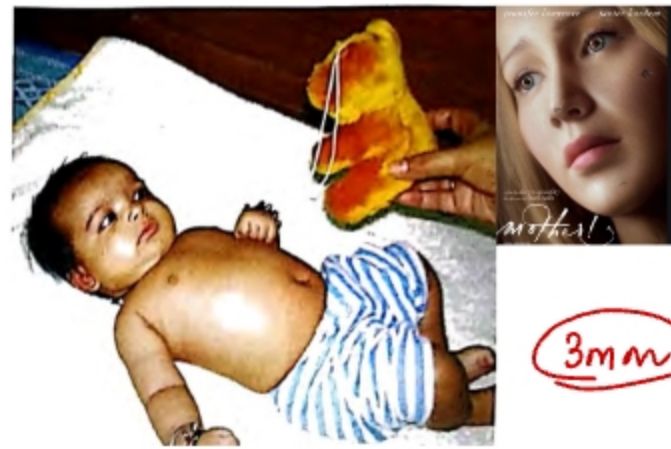
- **TrkA +**
- **<18months presentation**
- **Abundant lymphoid infiltrates**
- **Location in neck, thorax, pelvis**

Developmental milestones

1mon
↓
visual fixation

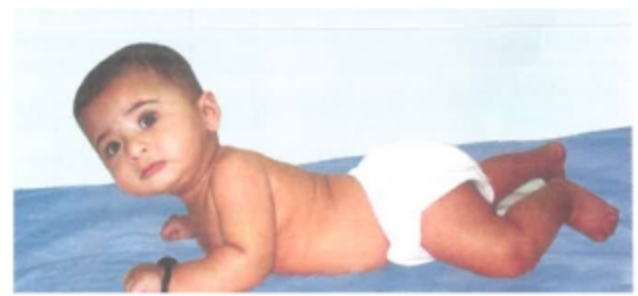


2mm
↓
social smile



3mm

- Palmar grasp disappears
- Recognize mother
- Hand regard (>20wks:abN)
- Cooing
- Neck holding
- Head above trunk in ventral suspension



4mon

- Bidextrous reach
- Binocular vision
- Mouthing
- ROFL (rolling laugh out loud)
- Pulls to sit; no head lag

6mon

Mirror play
Unidextrous, transfers
Monosyllables
Tripod sit & support △
Stranger anxiety
Listens to no
Sitting red flag: 10mon

9mm

Immature princer grasp
Object permanence
Bye-bye
"Bye"-syllables
Sits without support
Crawling

10mon

Pivots and cruises
Diagonal localization of sound
Stands with support
Creeps
Peek-a-boo
Standing red flag: 17mon

12mon

Mature princer grasp
Mouthing disappears
Comes when called
Stand without support
Throw ball
Casting
1-2 words

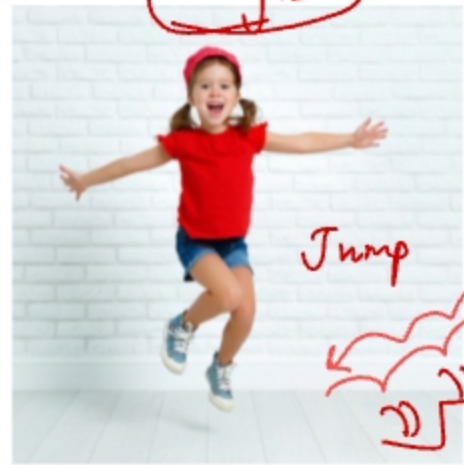
15mm

- Walk alone
- Jargon
- Imitates scribbling
- Turn 2-3 pages at a time
- 2 blocks tower
- Walking red flag: 18mon

18mm

- Domestic mimicry
- Explores drawer
- Unzips
- Runs and kicks ball
- Feeds with spoon
- Dry during day
- 8-10 words
- Tower of 3 cubes

2yrs



- 2 steps up and down
- Walk backwards
- 2 word sentences
- 50–100 words
- 2 objects
- Draw 2 lines
- Parallel play
- Tower-6 blocks (x 3)
- Asks for food and drink
- Names body parts
- Undresses completely
- Unscrew, door knobs
- Turn one page at a time

3yo

- Circle
- 1 step upwards
- Handedness
- 1-2 colours

- 9 tower (x 3)

- Dress + undress except buttons
- Name, gender, age

tricycle

Hopping

- 1 step downstairs
- Square, Cross
- Scissor
- Bridge with blocks
- Poem
- R-L discrimination
- Toilet alone

skip

- Triangle draw
- 3 step command
- Tie shoelaces
- Ask meaning of words
- Recognise 5 colours
- Gate with blocks
- Dress and undress without help

Age	Gross motor Milestone
3m	Neck holding
4m	Rolls over
6m	Sits in tripod position
9m	Sits without support, crawling
10m	Stand with support, creeps
12m	Stands without support
15m	Walks alone
18m	Runs
2y	Walks up and downstairs, 2 feet step
3y	Rides tricycle, alternate feet going upstairs
4y	Hops on one foot, alternate feet going downstairs

Age	Fine motor milestone
4m	Bidextrous approach
6m	Unidextrous approach
9m	Immature pincer grasp
12m	Mature pincer grasp
15m	Imitates scribbling, tower of 2 blocks, drinks from cup
18m	Scribbles, tower of 3 blocks
2y	Tower of 6 blocks, verticular and circular strokes, undresses, , feeds with spoon
3y	Tower of 9 blocks, copies circle, dresses
4y	Copies cross, bridge with blocks
5y	Copies triangle, gate with blocks

Age	Social Milestone
2m	Social smile
3m	Recognizes mother
6m	Stranger anxiety, inhibits to no
9m	Waves bye-bye, repeats activity when appreciated
12m	Comes when called, plays simple ball game
15m	Jargon, points to objects of interest
18m	Copies parents in task
2y	Asks for food, drink, toilet
3y	Shares toys, knows full name age gender
4y	Plays cooperatively in group, goes to toilet alone
5y	Helps in household tasks

Age	Language milestone
1m	Alerts to sound
3m	Coos
4m	Laugh loud
6m	Monosyllables
9m	Bisyllabes
12m	1-2 words with meaning
18m	8–10-word vocabulary
2y	2-3 word sentences, uses pronouns
3y	Asks question
4y	Sings song, tell stories
5yr	Asks meaning of words

Developmental quotient= $\frac{DA}{CA} \times 100$
Abnormal: < 70%
Global developmental delay: ≥ 2 domains
Preterm: 34wks – 40wks (6wks)
Screening: Denver
Goodenough-Harris
Trivandrum development
Phatak Baroda

Definitive: Bayley
Stanford Binet
Welscher Intelligence
Vineland adaptive

Intellectual disability	IQ	Disability %	Maximum functioning age
Mild	50–69	50%	12 years
Moderate	35–49	75%	9 years
Severe	20–34	90%	6 years
Profound	< 20	100%	3 years

Nocturnal enuresis
Twice a month in ≥ 5 **years**
Mx: Behavioral changes
↓
~~dry~~ Alarm therapy
↓
Pharmacotherapy : Desmopressin / Imipramine

NEONATOLOGY

LBW-<2.5kg
VLBW-<1.5kg
ELBW-<1kg
SGA-<10th %ile
AGA-10th-90th %ile
LGA->90th %ile

Physiological weight loss:
Term: 10% - required w/in 10days
Pre-term: 15% - regained w/in 15days

- Infant of diabetic mother
- Sotos syndrome
- Beckwidth Weidmann syndrome

MCC Of mortality:
Neonate – Prematurity > Asphyxia > Sepsis > Congenital anomalies
U5- Prematurity > Neonatal infection> Asphyxia > Pneumonia > Diarrhea

IUGR:<10th %ile weight (foetal) + abN doppler indices

	Symmetrical	Asymmetrical
Etiology	Congenital infections/ TORCH	Uteroplacental insufficiency
HC	Reduced	'Brain sparing'; Normal
AC	Reduced	Reduced
Ponderal index (g/cm3)	>2 - Normal	<2
Prognosis	Poor	Good

Expanded new Ballard score: 20-44 weeks

Neuromuscular Maturity

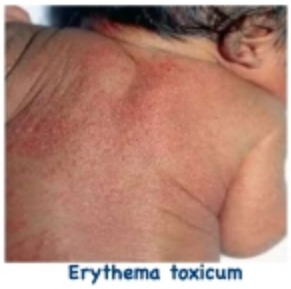
Score	-1	0	1	2	3	4	5
Posture							
Square window (wrist)	>90°	90°	60°	45°	30°	0°	
Arm recoil		180°	140°-180°	110°-140°	90°-110°	<90°	
Popliteal angle	180°	160°	140°	120°	100°	90°	<90°
Scarf sign							
Heel to ear							

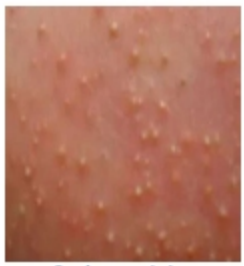
Physical Maturity

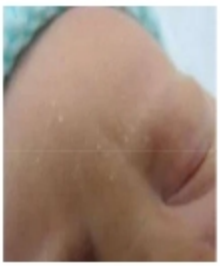
							Maturity Rating	
Skin	Sticky, friable, transparent	Gelatinous, red, translucent	Smooth, pink, visible veins	Superficial peeling and/or rash; few veins	Cracking, pale areas; rare veins	Parchment, deep cracking; no vessels	Leathery, cracked, wrinkled	Score / Weeks
Lanugo	None	Sparse	Abundant	Thinning	Bald areas	Mostly bald		-10 / 20
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		-5 / 22
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		0 / 24
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		5 / 26
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		10 / 28
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		15 / 30
								20 / 32
								25 / 34
								30 / 36
								35 / 38
								40 / 40
								45 / 42
								50 / 44

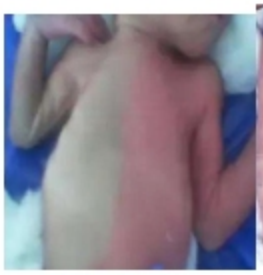
Max. score (@44 weeks): 50
Min. score (@20 weeks): -10

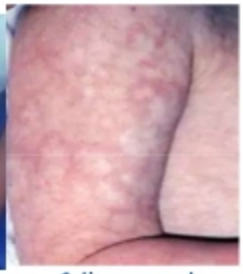
Benign newborn lesions

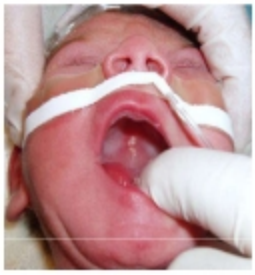

Erythema toxicum neonatorum
Eosinophilic aspirate

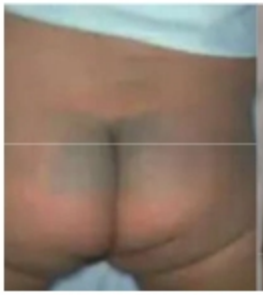

Benign pustular dermatosis
Neutrophilic aspirate

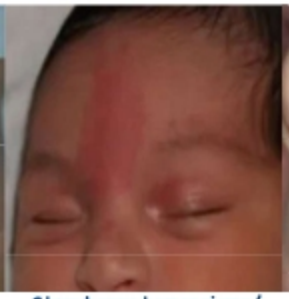

Milia
Obstruction of sweat gland ducts

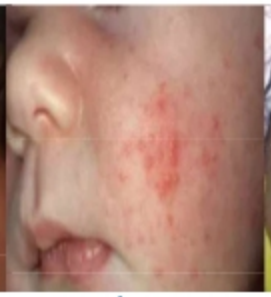

Harlequinn change

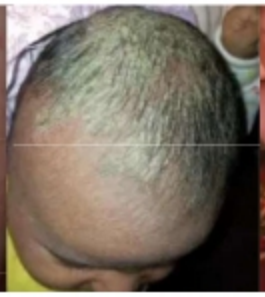

Cutis marmarota

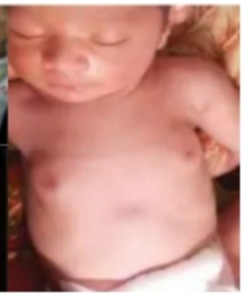

Epstein pearls (keratin)

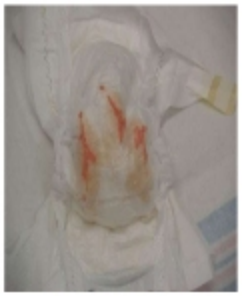

Mongolian spots
Dermal melanosis


Strawberry hemangioma/ Stork bite
Capillary hemangioma
Involutes by 1-2 years

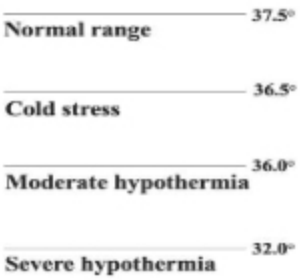

Acne

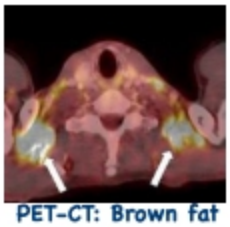

Cradle cap/Seborrheic dermatitis
r/o of LCH[+polydypsia/ polyuria + bony lesions]


Breast engorgement- Mastitis neonatorum


Pseudo-menstruation

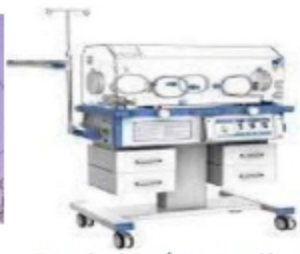
HYPOTHERMIA





PET-CT: Brown fat Axilla/Shoulder [Called Hibernoma]

Kangaroo Mother Care



Incubator (Convection)

Radiant warmer (Radiation)

- Axilla - 3 min
- Non-shivering thermiogenesis- brown fat uncoupler
- False +ve: PET

1. Kangaroo position (skin to skin) → prevent hypothermia (conduction)
2. Kangaroo nutrition (Exclusive bf)
3. Kangaroo support
4. Kangaroo discharge
   early neonatal sepsis

Indication: All stable LBW

min 1hr - as long as possible

Neonatal Hypoglycemia

Definition: Blood <40mg/dL/Plasma <45mg/dL

Asymptomatic: >20mg/dL - oral [BF] - >40mg/dL
└ <20mg/dL —>40mg/dL —> Glycaemic infusion

Symptomatic: iv dextrose bolus 2mL/kg - infusion [jitteriness; seizure; coma]

Features Differentiating Jitteriness from Seizures

1. Absence of eye deviation or fixed gaze, heart rate changes.
2. Rhythmic tremors (7-10Hz) with equal to and fro movements VS seizure is slower (1–2 Hz), with rapid and slow components.
3. Stimulus sensitive, precipitated by hunger, crying, or loud noise, and stopped by gentle restraint.

Neonatal sepsis

-MCC of neonatal sepsis- Acinetobacter > Klebsiella
-MCC of early onset sepsis (<72hrs)- GBS - S. agalactiae
-Most effective method for prevention- Hand washing (3min)
-Earliest C/F: Inadequate feeding
-Initial Ix: Sepsis screen -IOC: Culture
-R/F: Preterm, ROM >18hrs, PPV
Rx: Empirical: Ampicillin + Gentamicin +/- Vancomycin (after blood culture)

Sepsis Screen ≥2/5 +ve

- Leukopenia (TLC < 5000)
- Neutropenia (ANC < 1800)
- Immature to total neutrophil (I/T) ratio >0.2
- Micro-ESR > 15mm 1st hour
- CRP + ve

Normal Urine: ≤24hrs
Meconium: ≤48hrs
(biliverdin)

NEONATAL JAUNDICE

PATHOLOGICAL:

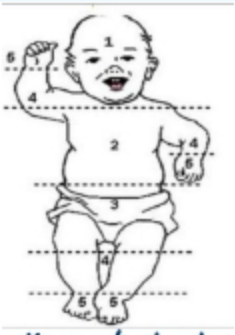
- Appears <24hrs

Causes: ABO incompatibility(O-Mother)/G6PD/HS

- Jaundice persisting > 14 days Prolonged

Causes: Hypothyroid/Cephalhematoma/BM jaundice

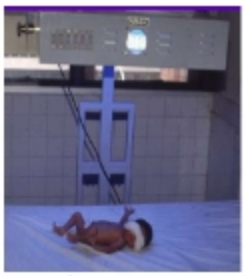
- Increase of bilirubin >5mg/dl/day
- Serum bilirubin>15mg/dl
- Conjugated Bilirubin >1mg/dl



Kramer's chart

Area of Body	Approximate Serum Bilirubin Level
Face	4–6 mg/dL
Chest	8–10 mg/dL
Abdomen	10-12 mg/dL
Limbs	12-14 mg/dL
Palms, soles	>15 mg/dL

TCB –> total serum bilirubin



S/E: Bronze baby/gonadal & retinal defects/ diarrhoea

Mechanism:

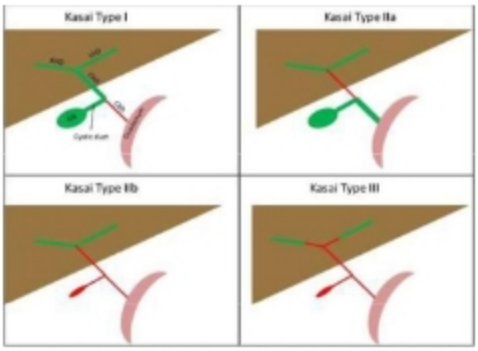
Structural isomerization: most imp.[Bil – Lumirubin]
Photo isomerization: Z –> E
Photo-oxidation least imp
-Distance: 30-45cm
-Wavelength: 450nm
-Type of lamp: LED
-Irradiance using flux meter:30 uW/cm2/nm

	Phototherapy	Exchange transfusion
24-48hrs	>15	>20
48-72hrs	>18	>25
>72hrs	>20	>25

*3% Breastfed babies develop BM jaundice

UNCONJUGATED

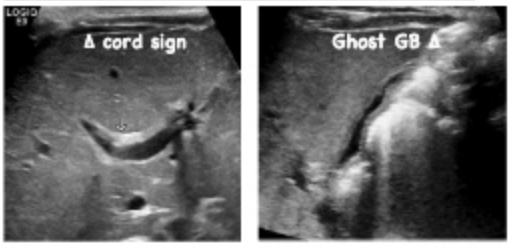
Criggler Najar syndrome AR	Gilbert syndrome AR	BREAST MILK JAUNDICE*	BREASTFEEDING JAUNDICE
UDP-GT severely deficient Kernicterus Type 1: Absent Type 2: deficiency (Rx w/ Phenobarb)	UDP-GT mildly deficient Stress/ fever	Prolonged jaundice Pregnanediol UDP-GT Rx: PhotoTh. Continue Bf	First week Feeding inadequate Rx: correct feeds Supplement: breast milk bank Formula

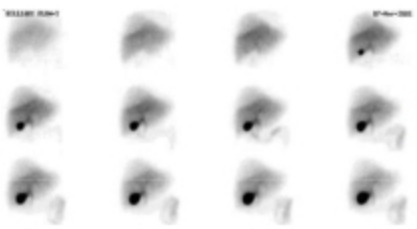


Type 3: MC

CONJUGATED: Direct bilirubin >2mg/dl

Dubin-Johnson AR	Rotor AR	EHBA	Neonatal hepatitis
MRP-2 Black liver Adrenaline metabolites	No black liver	Periductal fibrosis and proliferation IOC: Fasting USG > Tc99 HIDA > Biopsy > Intra-op cholangiography TOC: Kasai proc: HJ(<60d, x cirrhosis) – if fails/CI -> Liver transplant	Giant cells, lymphocytes



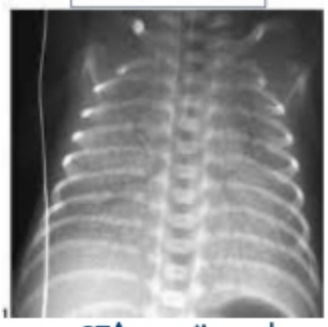


Tc99 HIDA scan

Neonatal respiratory distress

Hyaline membrane disease

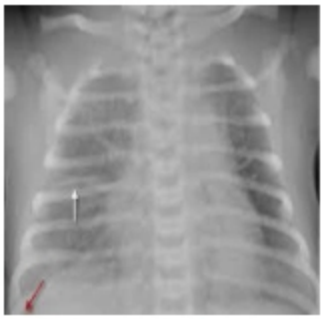
PRETERM



ST↑ compliance↓
Whiteout lung; Air bronchogram; Underinflated

TTNB

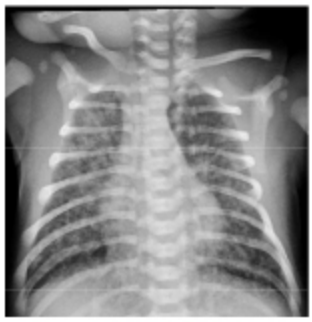
TERM / LSCS



Fluid in fissure/CP angle

Meconium Aspiration Syndrome (MAS)

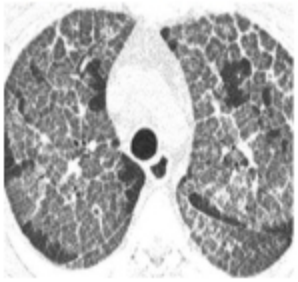
POST-TERM MSL



Hyperinflated lung
BV markings
Streaky opacities

Pulmonary Alveolar Proteinosis (PAP)

H/o sibling death
SP-B -macrophages



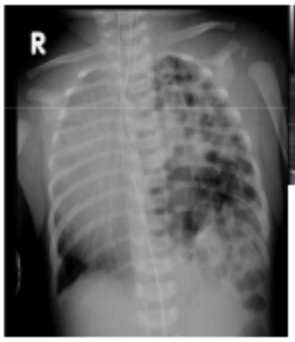
Crazy paving
RX: Whole lung lavage

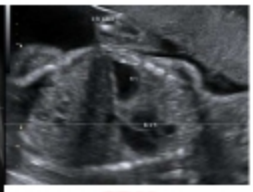
Surfactant- Type 2 pneumocytes
Synthesis begins- 20wks
Amniotic fluid- 28wks
Mature lungs- 34wks
Lecithin: Sphingomyelin ratio >2:1
(Dipalmitoyl phosphatidylcholine)
Nile blue sulfatase test → orange - mature lung

Shake test
[+ 95% ethanol]

Rx of HMD: CPAP/Mechanical ventilation- if fiO2 ≥40%
INSURE: Intubate → Surfactant → Extubate
LISA / MIST: Less Invasive Surfactant Administration; Minimally Invasive Surfactant Treatment
Prophylactic surfactant: ≤28wks POG
Failure to "wean off": Bronchopulmonary dysplasia
1-7 days Preterm: Apnoea of prematurity[≥20s + cyanosis/bradycardia]
Rx: Caffeine

Congenital pulmonary malformations



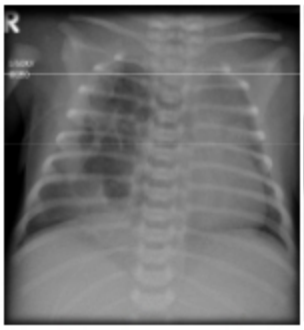


CDH

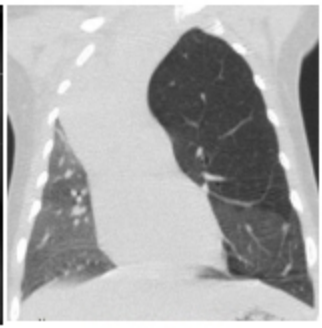
Absent pleuro-peritoneal membrane
1) Morgagni: Rt. & Ant.; adults; incidental
2) Bochdalek: MC; Lt. & Post.; Neonatal respiratory distress; Scaphoid abdomen → BMV C/I
Most important prognostic factor: Pulmonary HYPOPLASIA
1st: ETT → NGT f/b CXR f/b Surgery

CDH

Prognostic Lung:Head ratio
<1 = poor prognosis
LHR <1 → EXIT procedure

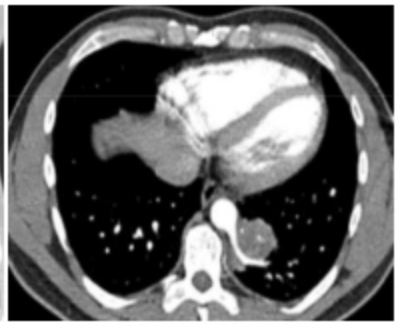


CPAM[Congenital Pulmonary Airway Malformation]



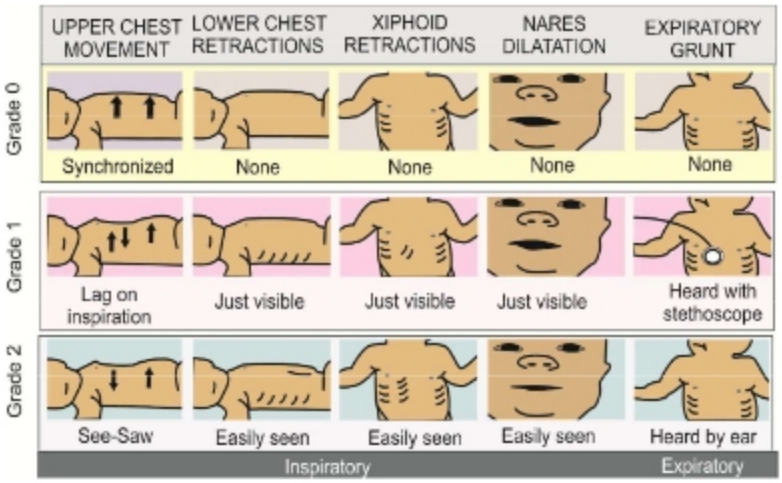
CLO/CLH

Congenital Lobar Overinflation/ Hyperinflation
MC: LUL



SEQUESTRATION

MC: LLL
Systemic arterial supply from Aorta +nt
r/o Recurrent pneumonia
Extra-lobar: systemic venous
Intra-lobar: pulmonary venous



Silverman-Anderson staging
Preterm neonates

Score	0	1	2
Respirate Rate	<60	60-80	>80
Cyanosis	None	No cyanosis with oxygen	Cyanosis with oxygen
Retraction	None	Mild	Moderate to severe
Grunting	None	Audible with stethoscope	Audible without stethoscope
Air Entry	Good	Decreased	Barely Audible

Downe's staging
Term & Preterm neonates

APGAR

Doesn't guide resuscitation

APGAR Score	Score 2	Score 1	Score 0
Appearance	 Pink	 Extremities Blue	 Pale or Blue
Pulse	>100 bpm	<100 bpm	No pulse
Grimace	Cries and pulls away	Grimaces or weak cry	No response to stimulation
Activity	 Active movement	 Arms, legs flexed	 No movement
Respiratory effort	Strong cry	Slow, irregular	No breathing

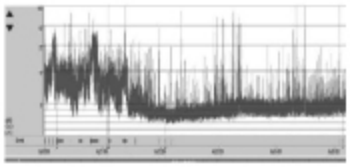
Prognostic score
Timing: 1, 5 min

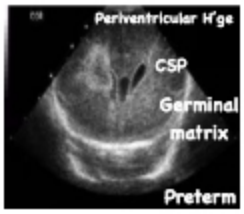
Severe asphyxia:
- APGAR: ≤3 @ 5min
- pH: <7.0
- CNS dysfunction:
- Hypotonia/seizures/ AbN Moro
- MODS

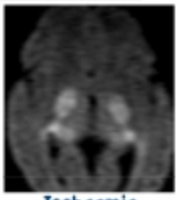
HIE

Neonatal seizures
MC TYPE- Subtle seizures[immature cortical network]
Best prognosis- Focal clonic type
Worst prognosis- Myoclonic type
MC CAUSE- HIE
CAUSES- ↑|↓ Na, ↓ Ca, Pyridoxine def., Hypoglycemia
DOC- Phenobarbitone -x-> Pyridoxine[B6]

BEDSIDE MONITOR- Integrated Amplitude electrode EEG
INITIAL IX- Transcranial USG - Ant.Fontanelle(Periventricular h'ge)
IOC- DWI(MRI)






Ischaemia (diffusion restriction +)

Patterns:
Cerebral palsy-SPASTIC type MC
Non-reversible, non-progressive
Term infant
- Parasagittal injury: Spastic quadriplegia
- Status marmoratus: Choreoathetoid (MC)

Preterm infant
- Periventricular leukomalacia → Spastic diplegia

Persistent primtive reflex
Cortical thumb
Commando crawl
Scissoring gait

Parameter	Stage 1 (Mild)	Stage 2 (Moderate)	Stage 3 (Severe)
Consciousness	Hyperalert	Lethargic	Coma
Activity	Normal	Decreased	Absent
Moro	Strong	Weak	Absent
Pupils	Mydriasis	Miosis	Variable
Heart rate	Tachycardia	Bradycardia	Variable
Seizures	None	Common	Uncommon
Prognosis	99% normal	80% normal	50% death 50% sequelae

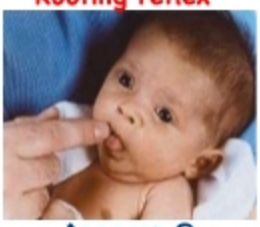
SARNAT & SARNAT STAGING

Neonatal reflexes

Palmar grasp

Appears @ 28wks
Disappears @ 3mon

Rooting reflex

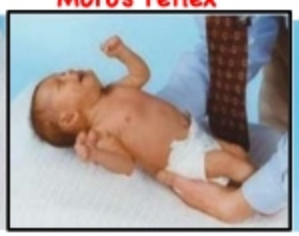


Appears @ 32wks
Disappears @ 1mon
1st to disappear

ASTNR

Appears @ 35wks
Disappears @ 6mon
I/L extension + C/L flexion

Moro's reflex



Appears @ 28-37wks
Disappears @ 6mon
Hands open + Shoulder abduct/ extend/ant. flexion of arm

Parachute reflex

Appears @ 8mon
Never disappears

Landau reflex
A: 3-4mon PN
D: 2yrs

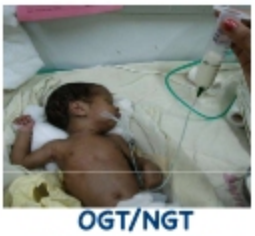
Persistent Moro's: Cerebral palsy
Exaggerated Moro's : HIE stage 1
Absent Moro's: HIE stage 2/3, Down's, Kernicterus
Asymmetric Moro's: Erb's palsy, #clavicle, #humerus

Appear after birth:
- STNR (I/L flexion)
- Landau's reflex
- Parachute reflex

STNR:
Appears @ 6-9 months
Disappears @ 12 months

Neonatal Feeding

Gestational age	Maturation of feeding skills	Initial feeding skills
<28 weeks	Inadequate sucking efforts Lack of gut motility	IV fluids
28-31 weeks	Sucking burst develop Lack of coordination between suck, swallow and breathing	Orogastric or nasogastric feeding
32-34 weeks	Coordination between breathing and swallowing begins	Spoon feeding
>34 weeks	Mature sucking pattern	Breastfeeding



OGT/NGT
Gavage feeding

Paladai feeding

Neonatal fluid requirement:
<1500g: 80ml/kg
>1500g: 60ml/kg
>1week: 150ml /kg

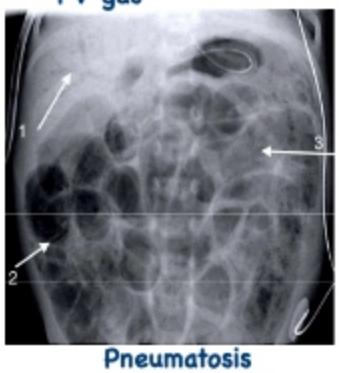
NEC

R/F: Preterm, Formula, PDA

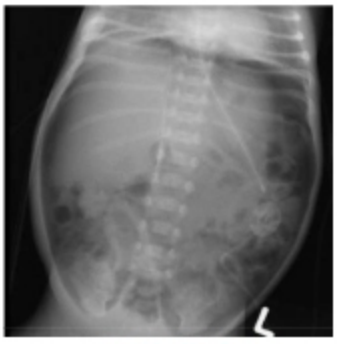
Stage	Systemic Signs	Treatment
IA BAT	Bradycardia, Apnea, Temperature instability	NPO Antibiotics Fluid resuscitation
IB	Grossly bloody stool	
IIA	Absent bowel sounds Pneumatosis intestinalis	
IIB	Metabolic acidosis, Thrombocytopenia PV gas	
IIIA	DIC	
IIIB	Pneumoperitoneum	Surgery

Modified Bell's staging

PV gas



Pneumatosis intestinalis



Football sign

TORCH infections

Toxoplasma

Hydrocephalus
Chorioretinitis Headlight in fog/Macular scar
Parenchymal calcification
Max transmission in T3

Microcephaly (<-2SD/<3rd %ile of HC) CMV
Periventricular calcification
MC long-term sequelae: SNHL
Most are asymptomatic (90%)
Urinary PCR test (or saliva)
Max r/o transmission from primary CMV

Zika virus

Microcephaly Aedes mosquitoe
Contractures
GM-WM calcification
TIM1 and TAM-XL

Varicella

Cicatricial skin rash
Limb hypoplasia
Max: 13-20 weeks
Neonatal varicella syndrome:
VZIG to infant: 5d before/ 2d after delivery
VZIg to pregnant female

Active genital herpes - indication of LSCS

Skin and eye lesions
Encephalitis
HSV-2

SNHL-MC finding Rubella
Cataract Nuclear pearly (MC)> Zonular/Lamellar
MC eye C/F: Salt & pepper retinopathy
PDA > PS
Expanded rubella: DM, renal diseases
Max transmission in in 1st trimester
Least r/o perinatal transmission

Non-immune hydrops
PRCA
Parvovirus B19

Growth

Weight with age:
Birth - x
20-40g/day x 3months
400g/month till 1 year
5mon - 2x
1yr - 3x
2yr - 4x
3yr - 5x
5yr - 6x
7yr - 7x
10yr - 10x

Height with age:
Birth 50cm
3mon 60cm
1yr 75cm (50%)
2yr 90cm (1/2 adult)
4yr 100cm
6cm / yr till 12yrs
(150cm)

Infantometer < 2yrs

US:LS :
Birth 1.8:1
3yr 1.3:1
7yr 1:1
>10yrs: 0.9:1

HC
Birth-32-35cm
1st 3month: 2cm/mon
Next 3month: 1cm/mon
Next 6month: 0.5cm/mon
Next 2 yrs: 0.2cm/mon
>2cm/month abN
12yrs: 52cm

Shaker's tape

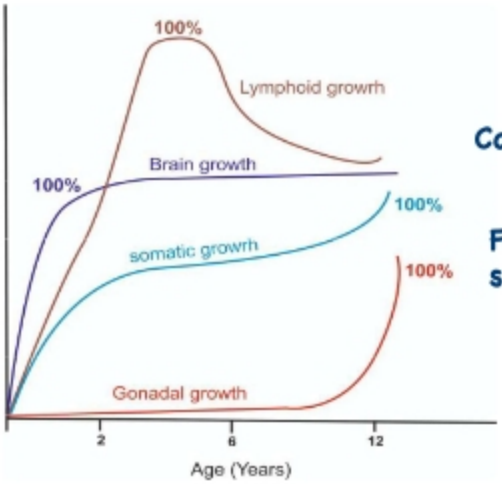
MUAC: age independent; 6m-5yrs

HC > CC by 2.5cm at birth
At 9-12mon: HC=CC - 47cm
>1yr: CC > HC

Surrogate marker of height: Arm span
Arm span < length by 2.5cm at birth
Equal at 11yrs
Arm span > length by 1cm after that
Mid-parental height: $\frac{\text{Mother+Father}}{2}$ +/- 6.5

Proportionate short stature: GH↓
Dispoportionate short stature-Short trunk
SED, MPS, Pott spine, Alagille Sx
Dispoportionate short stature-Short limb
Rickets, Achondroplasia, OI, Congenital hypothyroidism

Allagile syndrome: Hemivertebrae + Angular facies + Congenital hepatic fibrosis



SHORT STATURE: < -2SD/3rd %ile

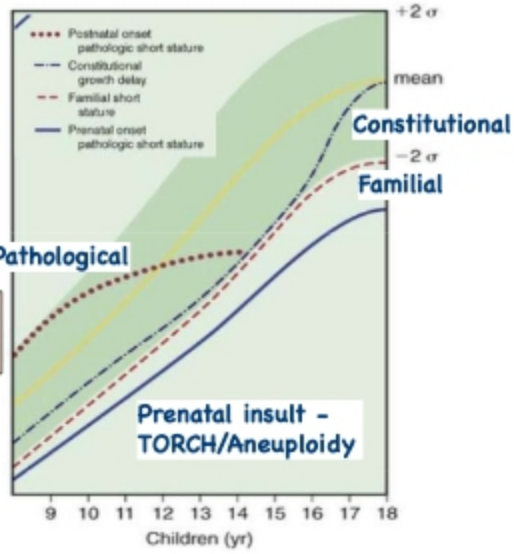
Bone Age < Chronological Age

Constitutional delay[Puberty delayed]

Bone = Chronological Age

Familial short stature[Puberty normal] **HypoThy GH↓**

Height velocity in CGD/Familial SS: Normal

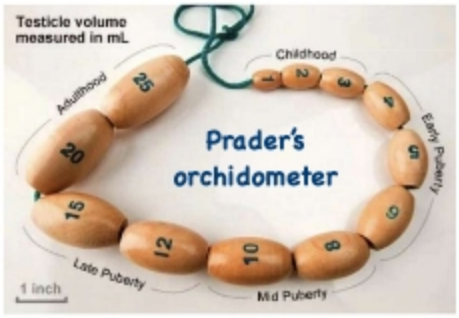


Puberty: TPM

FEMALES:	MALES:
Thelarche	Testicular enlargement
↓	↓
Pubarche	Penile enlargement
↓ — Growth spurt — ↓	
Menarche	Male voice/hair

Failure to thrive (FTT)
Descriptive term in <5yrs
Weight below the 3rd or 5th centile, failure to gain weight over time, or a change in the rate of growth such that weight for age or weight for length/height has crossed two major centiles (e.g., 50th to 10th) over a period of time

Tanner Stage	Male Genital Appearance	Female / Male Pubic Hair Appearance	Breast Appearance
Stage 1	Testicular volume < 3 mL	No pubic hair	Elevation of papilla only
Stage 2	Change in texture to scrotal skin	Sparse growth along the labia / base of penis	Breast bud stage
Stage 3	Increase in size of penis and testes	Darker, coarser, more curled hair	Enlargement of breast and areola
Stage 4	Further enlargement with development of glans penis	Adult-type hair over a smaller area	Projection of the areola and papilla
Stage 5	Adult size and shape	Spread to the medial surface of the thighs	Recession of the areola, projection of papilla only



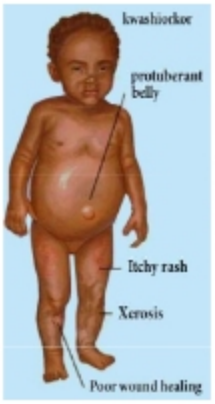
Five Components of Nurturing Child Care
1. Good health
2. Adequate nutrition
3. Responsive caregiving
4. Security and safety
5. Opportunities for early learning

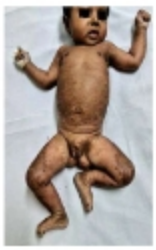
MALNUTRITION

Indicator	Parameter (<-2SD)	Interpretation
Stunting	Low height for age	Chronic malnutrition
Wasting	Low weight for height	Acute malnutrition
Under weight	Low weight for age	Acute on Chronic

Severe acute malnutrition (SAM):
6-59 months of age

- Weight-for-height below – 3 SD of the median
- Visible severe wasting
- Presence of bipedal edema
- Mid-upper arm circumference below 11.5cm





Kwashiorkor	Marasmus
Deficient of protein Serum albumin: <3g/dL	Deficient of proteins and calories Serum albumin: >3g/dL
Subcutaneous fat preserved Triceps skin fold: >50th %ile	Subcutaneous fat not preserved Muscle wasting Triceps skin fold: <3rd %ile
Oedema Flaky paint dermatitis Flag sign Fatty liver	Oedema absent Loose, wrinkled skin Simian facies
Lethargic	Alert and irritable
Poor appetite	Voracious feeder

Management of SAM

Poor appetite / Edema ++ /Medical complications

HOSPITAL Mx-
Sugar: hypoglycaemia- <54mg/dL - i.v. Dextrose bolus f/b infusion
Hypothermia
Infections
Electrolyte- Hypo-K/PO4
Dehydrations
Deficiency

Dehydration in SAM best assessed by: Urine output

Stabilisation: 0-7 days
F-75 (75 kcal + 0.9 g protein)/100 mL
All micronutrients except iron
Vit. A/K/Cu/Zn/Folate

Rehabilitation: 2-6weeks
F-100 (100 kCal + 2.9 g protein)/100 mL
Add iron

Primary failure:
- Failure to regain apetite by day 4
- Failure to lose edema by day 4
- Presence of edema on day 10
- Failure to gain at least 5g/kg/day by day 10

Breastfeeding

Constituent	Breast milk (gm/L)	Cow's milk (gm/L)
Proteins • Casein • Soluble proteins • Taurine, cysteine • DHA	11 4 7 + +	33 28 5 - -
Lactose	70	50
• Ca • P	0.33 0.15	1 1
Vitamins • C • D	 60mg 501U	 20mg 251U

Buffalo > Goat > Cow

Anti-infective:
TGF-B, Lactoferrin, IgA, Bifidus factor, Bile stimulated lipase
Colostrum: <3d –> ↑Ig –> transitional –> mature milk
Foremilk: water, protein, carbs, vitamins, minerals
Hindmilk: Calories <– Fat
Adequate feeding:
Sleep-2-3hrs Urine-6-8 times/d Gaining weight

Breast milk deficient in: vit. D (400IU at birth), vit. K(1mg), Fe (≥6m)
Max Breast milk output: 6mon
Expressed Breast milk: Room temperature 6hrs
Refrigerator 24hrs Deep freezer 3-6mon
Complementary feeding: Acceptable, Feasible, Affordable, Sustainable, Safe

Age	Quantity of feeds [250mL]	Frequency of feeds
6m-1yr	½ – 1 katori	3 times /day
> 1 year	1 – 1½ katori	5 times /day

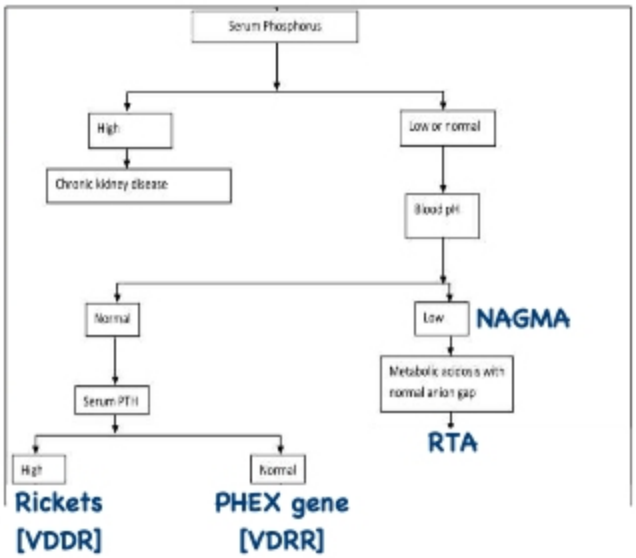
Adequate positioning:
Body well supported
Occiput, shoulder, buttocks in straight line
Entire baby turned towards mother
Abdomen touch-baby-mother

Adequate attachment:
Mouth wide open
Only small part upper areola visible
Lower lip everted
Chin touch mother's breast

Approach to rickets

Feature	Nutritional Rickets	Vit D–Dependent Type I (AR)	Vit D–Dependent Type II (AR)	X-linked Hypophosphat Rickets (XLD)	CKD
Defect	Vit. D ↓	1α-OH xx	End organ resistance	FGF-23 ↑ (PHEX)	1α-OH ⊖
Ca	↓	↓	↓	N	↓
PO_4	↓	↓	↓	↓	↑
PTH	↑	↑	↑	↑ – N	↑
$1,25(OH)_2D_3$	↓	↓	↑↑	N	↓
Response to Vit D	++	Calcitriol	↑doses of calcitriol	XX Rx – Burosumab	Vit. D Ca PO4 binders

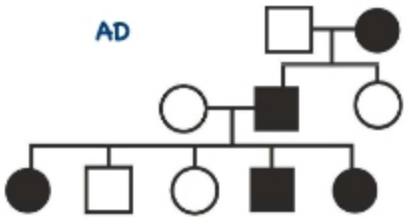
Rx: Vit.D
2000 IU
x 12wks

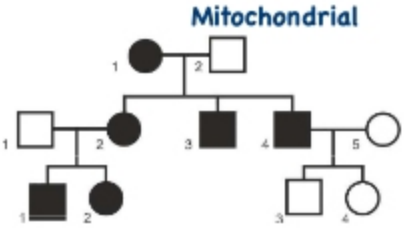


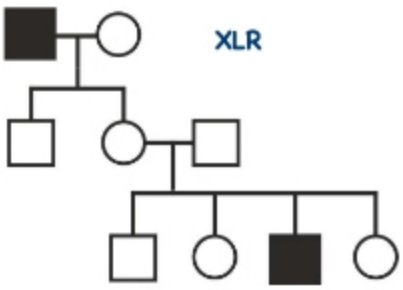
Tumour induced osteomalacia: FGF23 – Hemangiopericytoma & Hemangioma

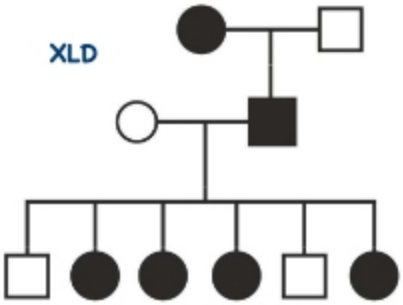
Pedigree Analysis

	STEPWISE:
AD (65%)	No gender pref All generations affected
AR (25%)	No gender pref Generations skipped
Mito-chondrial	Mother to ALL Heteroplasmy
Y - linked (Holandric)	Father to ALL sons
XLD	Father to all daughters Mother to 50% kids
XLR	Males affected, females carrier + Generations skipped
Gonadal - mosaicism	Surprise case!









INHERITANCE

AD

HS, VWD [except Type 3/2N]
Marfan, Achondroplasia/ EDS/ OI
AIP Noonan Sx
MEN, Neurocutaneous syndromes
SWS[Sporadic]; ATM[AR]

AR

All inborn errors of metabolism except
Sickle cell anemia, Thalassemia
CF Haemophilia C
Wilson, Hemochromatosis, Albinism, CAH

XLR

G6PD
DMD/ BMD
Hemophilia A / B
Wiscott Aldrich/ Bruton's/ CGD
Lesch Nyhan
Lowe Sx
Colour blindness
Menke's [ATP7A]
OTC deficiency
Barth Sx [Tafazzin gene]

Hunter's & Fabry's disease

XLD

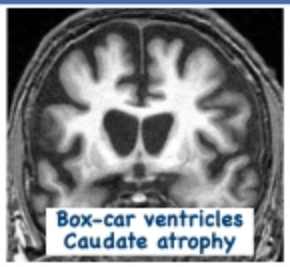
RP, Rett syndrome
Alport
Vit D resistant rickets
Incontinentia pigmentii

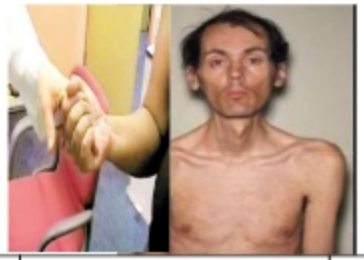
Mitochondrial

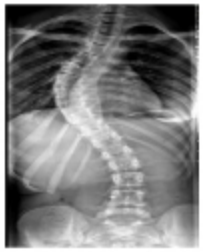
MELAS
MERRF
Kearns-Sayre/ CPEO
NARP
LHON
Leigh Sx
Pearson Sx

Pearson Sx: Pancreatitis + Sideroblastic anaemia

Trinucleotide repeat disorders



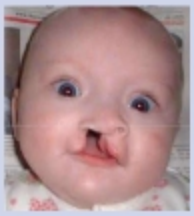


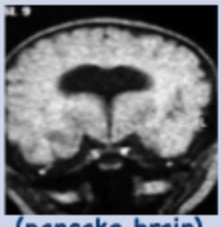


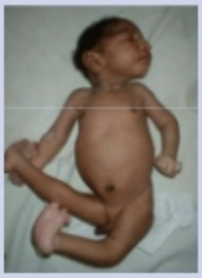
Diagnosis	Huntington chorea	Fragile X syndrome	Myotonic dystrophy	Friedrich ataxia
Repeat	CAG	CGG	CTG	GAA
Inheritance	AD	XLR/XLD	AD	AR
Chromosome	4	X	19	9
Gene	Huntingtin	FMR1	DMPK	Frataxin
Others	Sherman's paradox / Anticipation Caudate atrophy + low ACh/GABA + High Dopamine	MCC of Familial MR >200 repeats 55-200 repeats - ataxia + premature ovarian failure	Hatchet facies + Christmas tree cataract	Ataxia + Neuropathy + Scoliosis + Halux valgus Rx: OMAVEXOLONE

Genetic disorders- TRISOMY

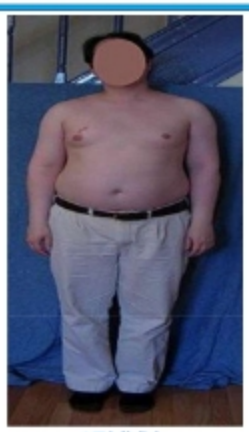
	DOWN (MC) (Chr.21)	PATAU (Chr.13)	EDWARD (2nd MC)
TONE	Reduced	Reduced	Increased (Chr.18)
HANDS	Simian crease/Cleinodactyly	Polydactyly	Clenched fist w/ overlapping finger
FEET	Sandal gap	Rocker bottom feet	Rocker bottom feet
EYES	Bruschfield spot/Epicanthal fold/Mongoloid slant	Hypotelorism/Cyclops eye	Microphthalmia
CVS	Endocardial cushion defect	VSD	VSD
MOUTH	Protruding tongue	Cleft lip/palate	Cleft lip/palate +/-
OTHERS	MC genetic cause of Low IQ GI: Duodenal atresia, Hirschprung disease AAD, Hypothyroid, Alzheimer's, ALL, AML-M CHL [Serous OM] Maternal Meiotic non-disjunction (95%) Robertsonian translocation-3% t[21;21] - 100% r/o recurrence	Not with maternal age PHPV d/t Persistent hyaloid artery Aplasia cutis Holoprosencephaly	Low IQ Maternal age Abdominal wall defects Horseshoe kidney




(pancake brain)



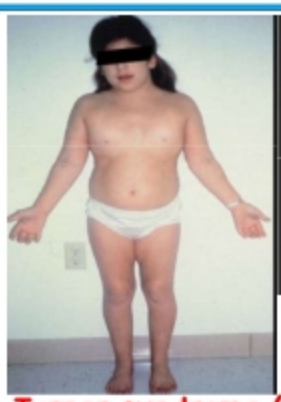
SYNDROMES

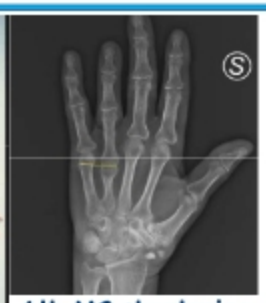


47XXY

Klinefelter syndrome
Non-disjunction of X
Paternal age
IQ: MR
Gonads: atrophy
Gynecomastia
Sparse hair

a/w Schizophrenia
FSH & LH high
Testosterone Low

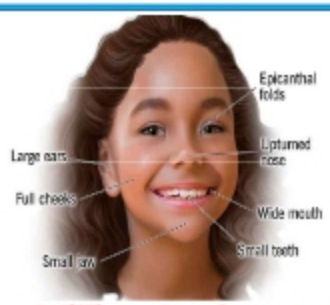




4th MC shortening

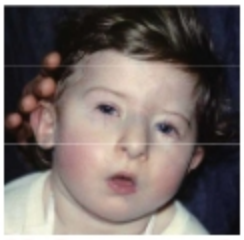
Turner syndrome (45 XO)

Mosaic: XO, XY: r/o gonadoblastoma
Lymphedema of feet and hands
Webbing of neck, Cystic hygroma
Shield chest, Inc carrying angle
Gonads: Streak ovaries
CVS: CoA < Bicuspid aorta
SNHL + Madelung deformity
IQ: Normal
NOONAN(AD): XX/XY + Phenotypic Turner + MR + Pulmonary stenosis

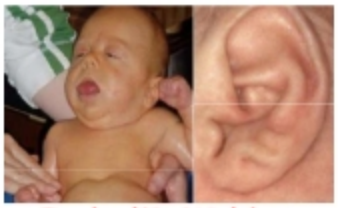


William Syndrome
μdeletion of Chr.7
Supravalvular AS +
Overfriendly + Elfian facies

Cri-du-chat syndrome
μdeletion of Chr.5 + shrill cry + anti-mongoloid slant



Di-George syndr.
μdeletion of Chr.22 + Hypocalcaemia + T-cell dysfunction

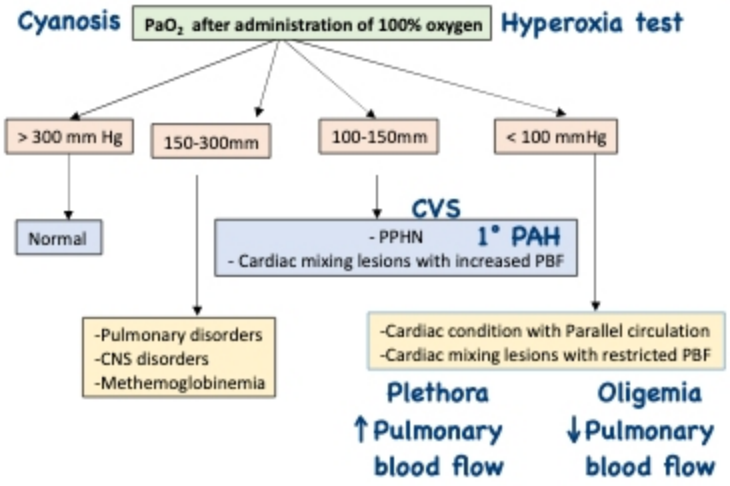


Beckwith-Weidmann syndr.
WT gene[Chr.11] defect
LGA + Macroglossia + Ear creases + Umbilical hernia + Hemihypertrophy

Congenital Heart Diseases-Approach

-MC overall: VSD
-MC to be affected by IE: VSD
-MC cyanotic HD: ToF
-MC cyanotic HD in neonates: TGA/TAPVC[infracardiac]
-MC cause of death in first week: HLHS

NADA'S CRITERIA	
MAJOR	**MINOR**
1. Systolic murmur Grade ≥ 3 2. Diastolic murmur 3. Cyanosis 4. Congestive Heart Failure	1. Systolic murmur ≤ Grade 2 2. Abnormal Second hear sound 3. Abnormal ECG 4. Abnormal Chest Xray 5. Abnormal Blood pressure



Acyanotic CHD

Acyanotic to cyanotic : EISENMENGER SYNDROME

PULMONARY PLETHORA

2nd heart sound wide and fixed split

ASD

RAD — Ostium secundum (MC)

LAD — Ostium primium

Pansystolic murmur
LAD
MC type: Membranous

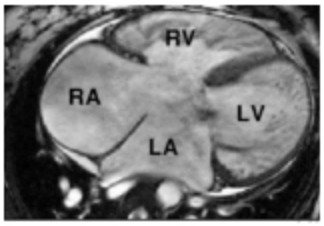
VSD

Continuous, machinery infraclavicular murmur
Differential cyanosis
PDA + Eisenmenger

PDA

Goose neck deformity
MC in downs

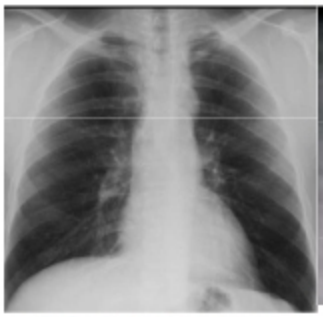
Endocardial cushion defect (AVSD)



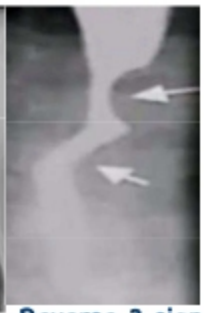
Normal PBF

Feeble femoral pulse
Brachio-femoral delay
Intermittent claudication
Hypertension

Coarctation of Aorta
Post ductal(MC)



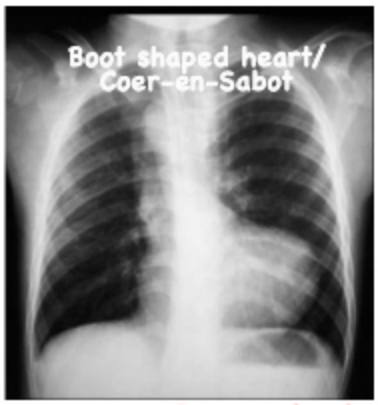
Inferior rib notching
Roesler's sign



Reverse 3 sign
(Ba swallow)

Cyanotic + Oligemia

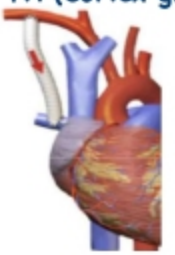
RAD
Pulmonary ejection systolic flow murmur
Single S2



Tetralogy of Fallot (ToF)

RVH[upturned apex] + Overriding of aorta + VSD (w/o HF) + Infundibular pulmonary stenosis[oligaemia]
Pentalogy: +ASD

Blalock-taussig: SCA-PA (Gortex graft)



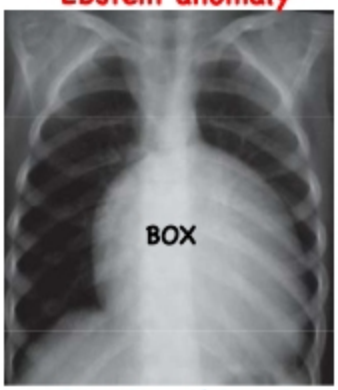
Potts: DA-PA
Waterston: AA-PA

Cyanotic spells Mx:

- Knee-chest/squatting position
- Morphine
- Oxygen
- β-blockers

WPW, RBBB, Himalayan P waves

Ebstein anomaly



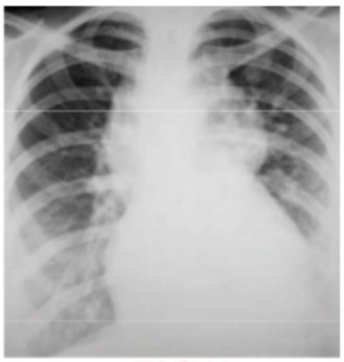
Box shaped heart
Maternal Lithium intake
Atrialisation of RV

LAD

Tricuspid atresia

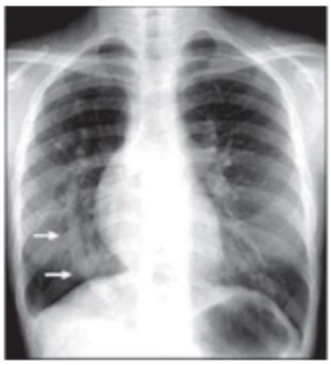
Cyanotic + Plethora

LARGE PEDICLE
2ND HS WIDE AND FIXED
Same saturation in all chambers



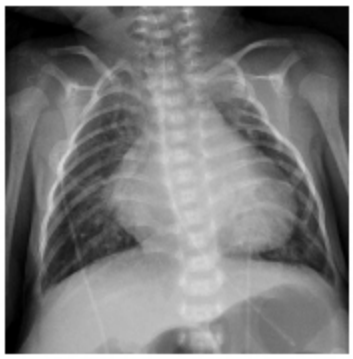
TAPVC
Figure of 8/Snowman/
Cottage loaf app.

- Type 1: Supracardiac (MC)
- Type 2: Cardiac
- Type 3: Infracardiac (IVC#
  - Poor prognosis



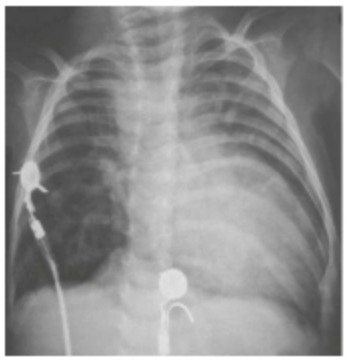
PAPVC
[Rt.Pulm.vein –> IVC]
Scimitar sign
Rt lung hypoplasia

SMALL PEDICLE



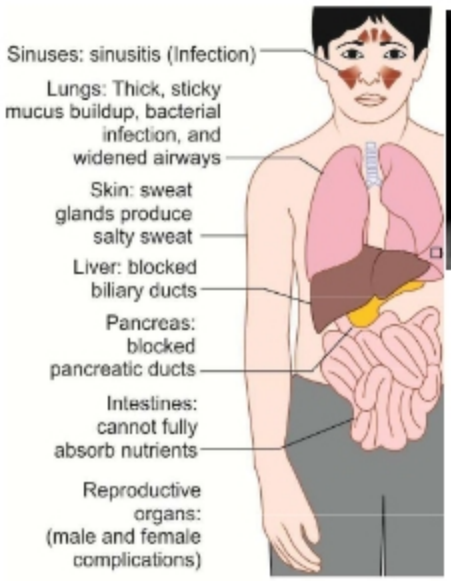
Transposition of Great Arteries (TGA)
Egg on a string app.
Septum dependent
PDA (DA-open) –>
Alprostadil (PGE1)
Atrial septostomy ->
Jatine's arterial switch

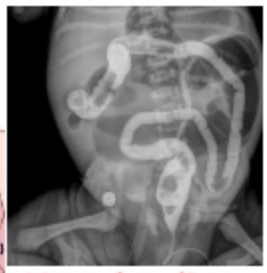
LARGE PEDICLE



Persistent Truncus Arteriosus (PTA)
Sitting duck sign

Cystic fibrosis





Meconium ileus w/ Microcolon
Dx+Rx:
Gastrograffin enema
Bishop-koop sx [if enema fails]

Cystic Fibrosis Finding	Biochemical Evidence of CFTR Dysfunction
• ≥1 Phenotypical finding • Positive neonatal screening: Immunoreactive trypsinogen • Positive family history	• Positive sweat chloride >60meq/l on 2 separate days • Positive Nasal potential difference • 2 mutations in CFTR

CF: Chromosome: 6(CFTR gene); AR
-MC mutation: ΔF508
-MC class of mutation: 2(Trafficking)
-Trikafta:
Elexacaftor + Tezacaftor (Correcter) + Ivacaftor (Potentiator)

	Sweat glands	Other ducts
Cl level	High	Low
Cl transport	Low	Low

0-12yrs: Malabsorption/ steatorrhea MC
>13yrs: Pneumonia MC
Azoospemia in 98%

Pneumonia:

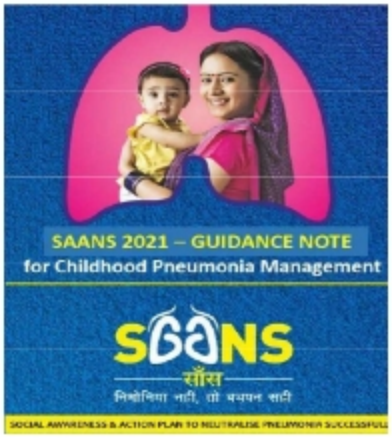
- <16yrs: S.aureus
- ≥16yrs: Pseudomonas
- Most sp: Burkholderia

Infertility:

- Male: Congenital B/L absent VD
- Female: Cervical mucous thickens

Pneumonia

IMNCI Category	Features	Management
No pneumonia	Fever, cough/cold	• Symptomatic • Follow-up after **5 days**
Pneumonia	• Fast breathing +/- chest indrawing	• **Oral amoxicillin × 5 days** • Follow-up after **2 days**
Severe pneumonia	• SpO_2 < 90% • Stridor in calm child • ≥1 danger sign: Lethargy, Not feeding, Convulsions, Cyanosis, unconscious, head nodding	• **Urgent referral** • Give **1st dose ampicillin + gentamicin im injection**



Fast breathing:

- 0-2m - ≥60
- 2-12m - ≥50
- 12m - ≥40

Acute Diarrhea

Parameters	No Dehydration	Some Dehydration	Severe Dehydration
Appearance	**Well, alert**	**Restless,** irritable	**Lethargic, unconscious**
Eyes	**Normal**	**Sunken**	**Very sunken**
Thirst	**Drinks normally, not thirsty**	**Thirsty, drinks eagerly**	**Drinks poorly or not able to drink**
Skin pinch	**Goes back quickly (<1 second)**	**Goes back slowly (1 second)**	**Goes back very slowly (2 seconds)**

ZINC: 14days
<6mon: 10mg
>6mon: 20mg

Oral fluids/ ORS

75ml/kg over 4 hours
ORS

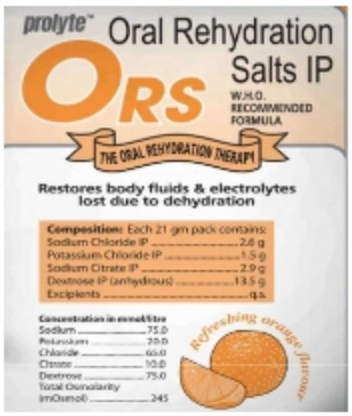
RL + 5% dextrose
100ml/kg
30ml/kg 70ml/kg

<1yr : 1hr 5 hrs
>1yr : 0.5hr 2.5hrs

FLUID COMPOSITION

Composition	ReSoMal (mmol/L) SAM	Standard ORS (mmol/L)	Reduced osmolarity ORS
Glucose	125	111	75
Sodium	45	90	75
Potassium	40	20	20
Chloride	70	80	65
Citrate	7	10	10
Magnesium	3	---	---
Zinc	0.3	---	---
Copper	0.045	---	---
Osmolarity (mOsm/L)	300	311	245

	Plasma*	0.9% NaCl	Hartmann's
Na^+ (mmol/l)	135–145	154	131
Cl^- (mmol/l)	95–105	154	111
$[Na^+]:[Cl^-]$ ratio	1.28–1.45:1	1:1	1.18:1
K^+ (mmol/l)	3.5–5.3	0	5
HCO_3^- / Bicarbonate precursor (mmol/l)	24–32	0	29 (lactate)
Ca^{2+} (mmol/l)	2.2–2.6	0	2
Mg^{2+} (mmol/l)	0.8–1.2	0	0
Glucose (mmol/l)	3.5–5.5	0	0
pH	7.35–7.45	4.5–7.0	5.0–7.0
Osmolarity (mOsm/l)	275–295	308	278



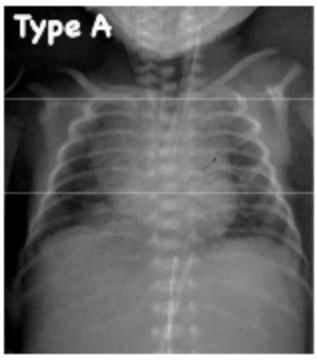
Fluid for maintenance: N + 5% dextrose
1st 10kg: 100ml/kg Next 10 kg: 50ml/kg >20kg: 20ml/kg

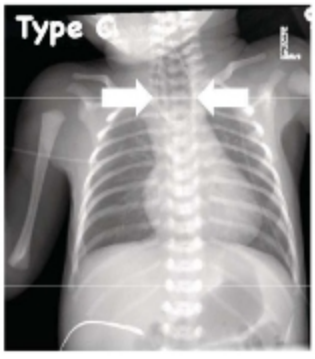
Holliday- Segar formula

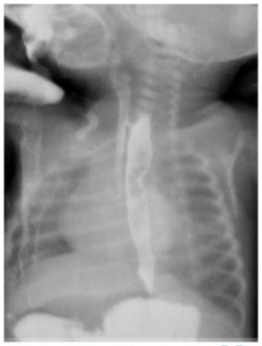
- NAGMA = Hyperchloremic acidosis –> hence NS preferred in vomiting
- RL: C/I in Cirrhosis [Lactate –> HCO3 - causes metabolic alkalosis]

GI emergencies

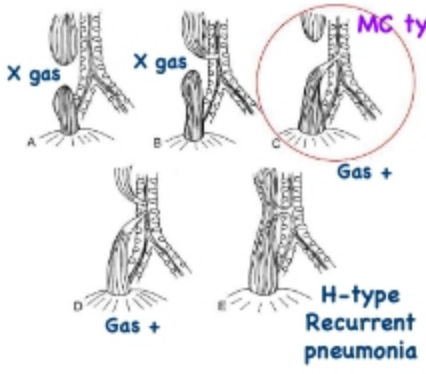
Newborn with drooling and difficulty feeding (TEF)





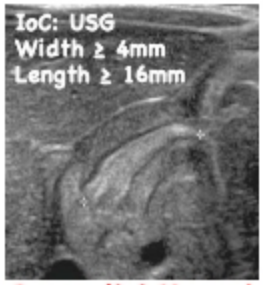


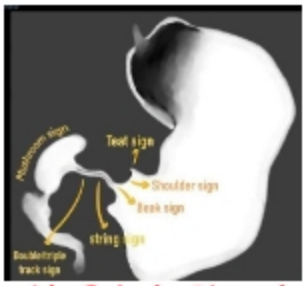
IoC: Gastrografin study



A/w VACTERL anomalies:
- Vertebral
- ARM
- Cardiac
- TEF
- Renal
- Limb[Radial ray]

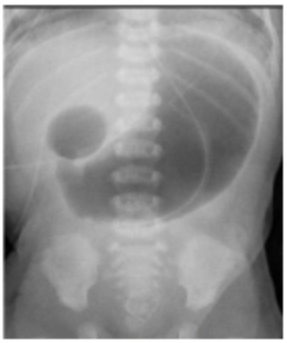
Non-bilious vomiting
Non-projectile- GERD
Sandifer Sx- posturing - arch back
Projectile- CHPS





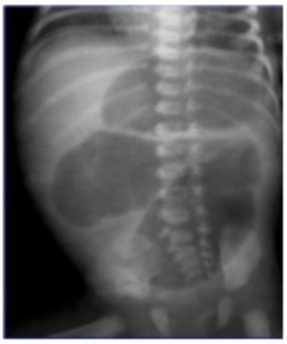
Congenital Hypertrophic Pyloric Stenosis [CHPS]
- olive shaped mass at LUQ
- L –> R peristalsis
- 4-6wks after birth
- a/w maternal macrolide cosumption
- Metabolic problem: Metabolic alkalosis + Hypochloremia + Hypokalaemia + Paradoxical aciduria
- Rx: Ramstead's pyloromyotomy/POEM

Billious vomiting



Double bubble sign

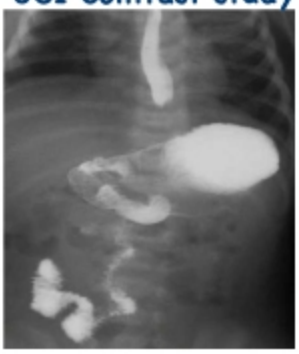
- Duodenal atresia
- Annular pancreas
- Ladd's band

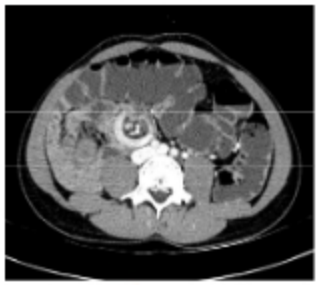


Triple bubble sign

Jejunal atresia

UGI Contrast study





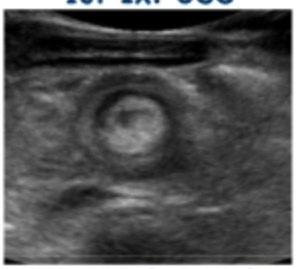
Corkscrew/Whirlpool sign

Midgut volvulus + Malrotation

Intussuception

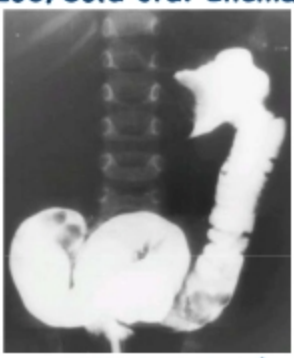
Pain + red currant jelly stools

1st Ix: USG



Target/Donut/ Sandwich/Pseudo-kidney sign

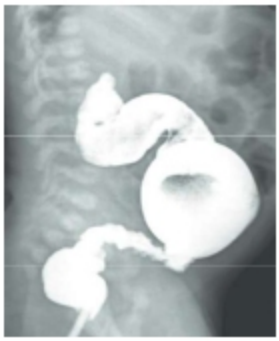
IoC/Gold std: Enema

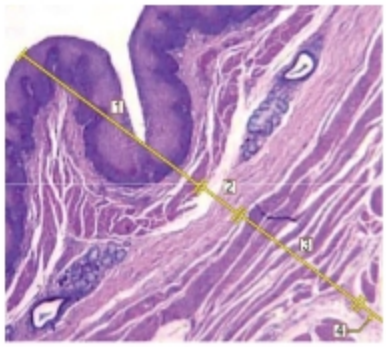


Coiled spring/ Claw sign

- Any enema[Ba,air,NS] can be used
- Rotavirus vaccine/Weaning are RFs
- Pathophys: Peyer's patch hypertrophy
- MC location: Ileo-colic
- Adult pathophys: d/t lead pt. - Polyp/Carcinoma/ Meckel's/HSP

Not passed meconium >48hrs after birth





Hirschprung's diease

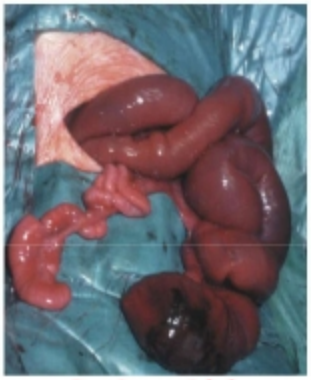
- Defect of Neural crest cell migration —> absent nerve plexus in rectum —> Absent peristalsis
- Sigmoid colon: MEGACOLON
- Rectum: AGANGLIONOSIS
- 1st Ix: Ba Enema
- IoC: Punch biopsy
- Rx: Pull through procedure - Swenson/Duhamel/Soave

At birth

Peritoneum
Amnion
Wharton's jelly
Minor:<5cm

Omphalocoele

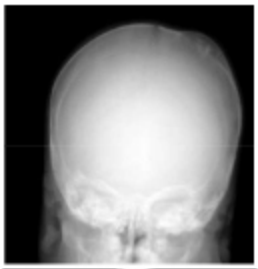
- Membrane +nt
- Apex of umbilicus
- High r/o congenital anomalies

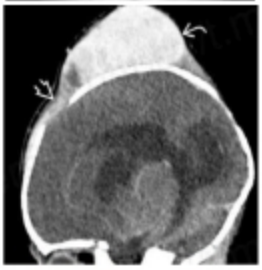


Gastroschisis

- Off midline
- High r/o bowel necrosis

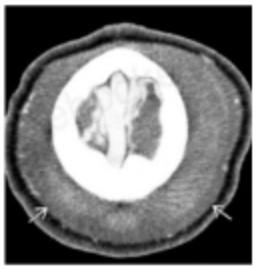
Skull pathologies



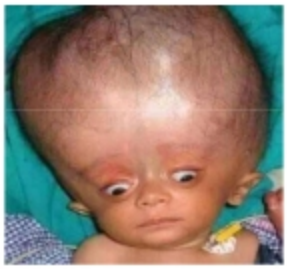


Cephalhematoma
Focal; Subperiosteal
May increase in size
Prolonged jaundice

Caput Succedenum
Focal; Edema
Max. size @ birth
Reassure parents

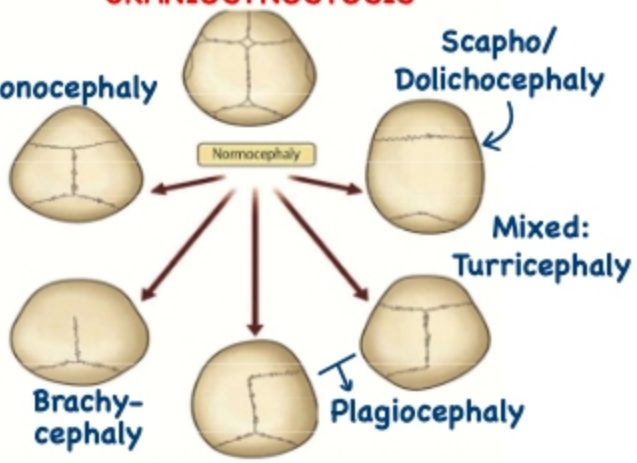


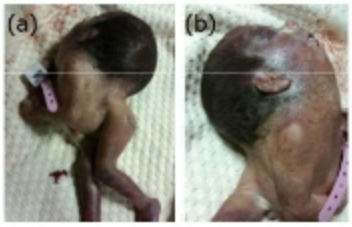
Subgaleal hematoma
Diffuse
r/o Hypovolemic shock



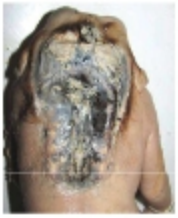
MCC: Aqueductal stenosis
Sunset sign Tectal plate compression
McEwen sign
Mx: VP shunt
IOC for shunt infection: Shunt tap

CRANIOSYNOSTOSIS

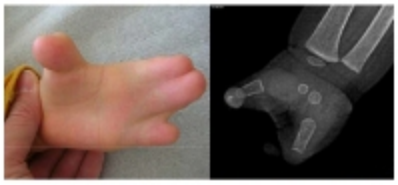




Iniencephaly



Craniospinal Rachischisis



- Apert syndrome: Syndactyly - Mitten hands
- Crouzon syndrome: Midfacial hypoplasia - FGFR2#
- Carpenter syndrome
- Pfieffer syndrome

Miscellaneous

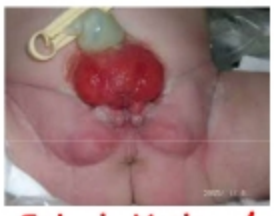
Febrile seizures: Fever (>100.4) + seizure in 6mon-6yrs
- Simple-GTCS, <15min, No recurrence in 24hrs
- Complex Poor prognosis

No long-term AED
If >5min: BZD DOC → Lorazepam/Rectal Diazepam/ i.n. Midazolam
High risk for recurrence – 30-50%
Risk factors:
MAJOR: <1yr, <24hr fever, 100-102 F
MINOR: Complex, Family history , Low Na, Male, Daycare

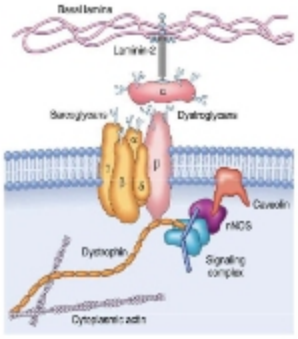
Schwartz formula

$$\text{eGFR} = \frac{\text{Length (cm)} \times k\,[0.41]}{\text{Serum creatinine (mg/dL)}}$$

Congenital hypothyroidism
Growth retardation, umbilical hernia, Large tongue, Epiphyses delayed
MCC: Dysgenesis > Dyshormonogenesis
Screening: TSH → ≥48hrs of birth → DBS/ Heel prick
Next: USG Thyroid/RAIU scan

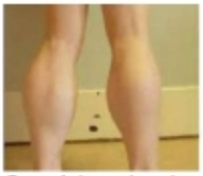


Ectopia Vesicae/ Exstrophy UB
High r/o: AdenoCa UB
Epispadias



Proximal weakness

GOWER SIGN

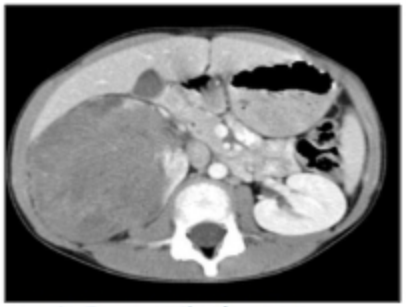


Pseudohypetrophy [Fatty replacement]

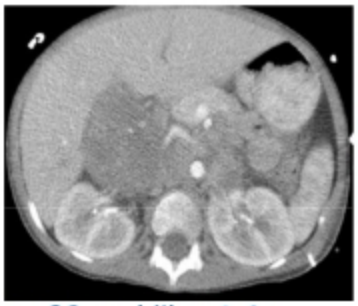
Muscular Dystrophy[XLR]

CPK very high, Dystrophin gene
Frameshift / Non-sense: Absent: Duchhene's
In-frame mutation: Reduced: Becker's
Cardiomyopathy, Low IQ
MCC of death: Resp.muscle weakness
Eteplirsen Exon 51
Casimersen Exon 45

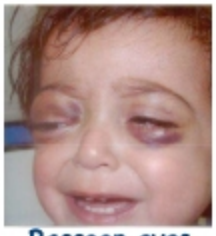
Wilms tumor VS Neuroblastoma



- 0-5yrs - Flank mass
- Invasion - RV/IVC
- Mets - MC in lungs

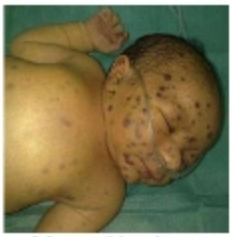


- RP - midline Ca2+
- encase vessels
- Mets - MC in Bones



Raccoon eyes
↓
Mets to orbit

Opsoclonus-Myoclonus



Skin - Blueberry muffin rash

Risk factors: **WILMS TUMOUR**
WT: Chr 11
11p13: WAGR
Denys Drash
11p15: Beckwith Weidmann
Horseshoe kidney
UDT, Hypospadias

WAGR: WT/Aniridia/Growth retardation & MR/GU anomalies
Denny's Drash syndrome: Mesangial sclerosis & GU anomalies

- Hypodiploidy
- N-myc amplification
- ALK amplification
- Loss of heterozygosity-1p,11q

Bad prognosis

- TrkA +
- <18months presentation
- Abundant lymphoid infiltrates
- Location in neck, thorax, pelvis

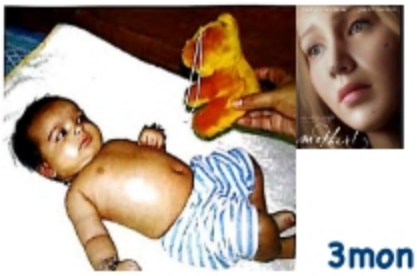
Good prognosis

NEUROBLASTOMA

Developmental milestones

1mon
Visual fixation

2mon
Social smile



3mon

Palmar grasp disappears
Recognize mother
Hand regard (>20wks:abN)
Cooing
Neck holding
Head above trunk in ventral suspension

4mon

Bidextrous reach
Binocular vision
Mouthing — Rolling
ROFL — Laugh out loud
Pulls to sit; no head lag

6mon

Mirror play
Unidextrous, transfers
Monosyllables
Tripod[sit w/ support]
Stranger anxiety
Listens to no
Sitting red flag: 10mon

9mon

Immature princer grasp
Object permanence
Bye-bye
"Bye"-syllables
Sits without support
Crawling

10mon

Pivots and cruises
Diagonal localization of sound
Stands with support
Creeps
Peek-a-boo
Standing red flag: 17mon

12mon

Mature princer grasp
Mouthing disappears
Comes when called
Stand without support
Throw ball
Casting
1-2 words

15mon

Walk alone
Jargon
Imitates scribbling
Turn 2-3 pages at a time
2 blocks tower
Walking red flag: 18mon

18mon

Domestic mimicry
Explores drawer
Unzips
Runs and kicks ball
Feeds with spoon
Dry during day
8-10 words
Tower of 3 cubes

2yrs

2 steps up and down
Walk backwards
2 word sentences
50-100 words
2 objects
Draw 2 lines
Parallel play
Tower-6 blocks
Asks for food and drink
Names body parts
Undresses completely
Unscrew, door knobs
Turn one page at a time

3yrs

Circle
1 step upwards
Handedness
1-2 colours

9 tower

Dress + undress except buttons
Name, gender, age

4yrs

Hopping

1 step downstairs Square, Cross Scissor Bridge with blocks Poem R-L discrimination Toilet alone

5yrs

Skip

Triangle draw 3 step command Tie shoelaces Ask meaning of words Recognise 5 colours Gate with blocks Dress and undress without help

Age	Gross motor Milestone
3m	Neck holding
4m	Rolls over
6m	Sits in tripod position
9m	Sits without support, crawling
10m	Stand with support, creeps
12m	Stands without support
15m	Walks alone
18m	Runs
2y	Walks up and downstairs, 2 feet step
3y	Rides tricycle, alternate feet going upstairs
4y	Hops on one foot, alternate feet going downstairs

Age	Fine motor milestone
4m	Bidextrous approach
6m	Unidextrous approach
9m	Immature pincer grasp
12m	Mature pincer grasp
15m	Imitates scribbling, tower of 2 blocks, drinks from cup
18m	Scribbles, tower of 3 blocks
2y	Tower of 6 blocks, verticular and circular strokes, undresses, , feeds with spoon
3y	Tower of 9 blocks, copies circle, dresses
4y	Copies cross, bridge with blocks
5y	Copies triangle, gate with blocks

Age	Social Milestone
2m	Social smile
3m	Recognizes mother
4m	Stranger anxiety, inhibits to no
6m	Waves bye-bye, repeats activity when appreciated
9m	Comes when called, plays simple ball game
12m	Jargon, points to objects of interest
18m	Copies parents in task
2y	Asks for food, drink, toilet
3y	Shares toys, knows full name age gender
4y	Plays cooperatively in group, goes to toilet alone
5y	Helps in household tasks

Age	Language milestone
1m	Alerts to sound
3m	Coos
4m	Laugh loud
6m	Monosyllables
9m	Bisyllabes
12m	1-2 words with meaning
18m	8–10-word vocabulary
2y	2-3 word sentences, uses pronouns
3y	Asks question
4y	Sings song, tell stories
5y	Asks meaning of words

Developmental quotient = $\frac{DA}{CA}$ X100
Abnormal: <70%
Global developmental delay: ≥2 domains
Preterm: 34wks – 40wks
Screening: Denver
Goodenough-Harris
Trivandrum development
Phatak Baroda

Definitive: Bayley
Stanford Binet
Welscher Intelligence
Vineland adaptive

Intellectual disability	IQ	Disability %	Maximum functioning age
Mild	50–69	50%	12 years
Moderate	35–49	75%	9 years
Severe	20–34	90%	6 years
Profound	< 20	100%	3 years

Nocturnal enuresis
Twice a month in ≥ 5 years
Mx: Behavioural changes
↓
Alarm Therapy
↓
Pharmacotherapy:
Desmopressin/Imipramine