APGAR SCORES EXPLAINED

	Indicator	0 Points	1 Point	2 Points
A	Appearance (skin color)	Blue; Pale	Pink Body; Blue Extremities	Pink
P	Pulse	Absent	Below 100 bpm	Over 100 bpm
G	Grimace (reflex irritability)	Floppy	Minimal Response to Stimulation	Prompt Response to Stimulation
A	Activity (muscle tone)	Absent	Flexed Arms and Legs	Active
R	Respiration efforts	Absent	Slow and Irregular	Vigorous Cry

APGAR Score is an objective method of evaluating the newborn's condition using 5 parameter's.

- It is performed at 1 minute & 5 minutes after birth.

Note: Apgar score is not used to guide resuscitation.

	Rotor syndrome	Dubin Johnson syndrome
Inheritance	Autosomal recessive	Autosomal recessive
Defect	Organic anion transporting polypeptide defect. (OATP1B1,OATP1B3)	Multi drug resistance protein (MRP)2 mutation
Kernicterus	Absent	Absent
Biopsy	Normal	Black pigmentation
Total Urinary coproporphyrin	Elevated	Normal
Urinary coproporphyrin I	<70%	>80%
Bromsulphalein clearance test	Slow clearance	No/Delayed clearance
Gall bladder on oral cholangiography	Visualized	Not visualized
Cholestasis	No	No
Serum bile acids	Normal	Normal
Jaundice	Mild (TB<4mg/dl)	Mild (TB<4mg/dl)

Congenital cytomegalovirus infection:

- m/c TORCH infection
- Risk of Transmission of CMV from mother to baby is high in 3rd trimester

1. Congenital / intrauterine transmission:

- 3rd trimester
- Around 90% babies asymptomatic
- <5% babies affected → Congenital cytomegalic inclusion disease.

Q. m/c long term sequealae: Sensorineural hearing loss.

Q. Δsis: PCR Testing

(test to be done within 2wks after birth as 2wks intrauterine & post natal infections can't be distinguished)

- Sample used → Urine > blood > Saliva (best)
- IgM test

CMV Triad

- Microcephaly
- Chorioretinitis
- intracranial calcification

↓

Periventricular / around the ventricles.

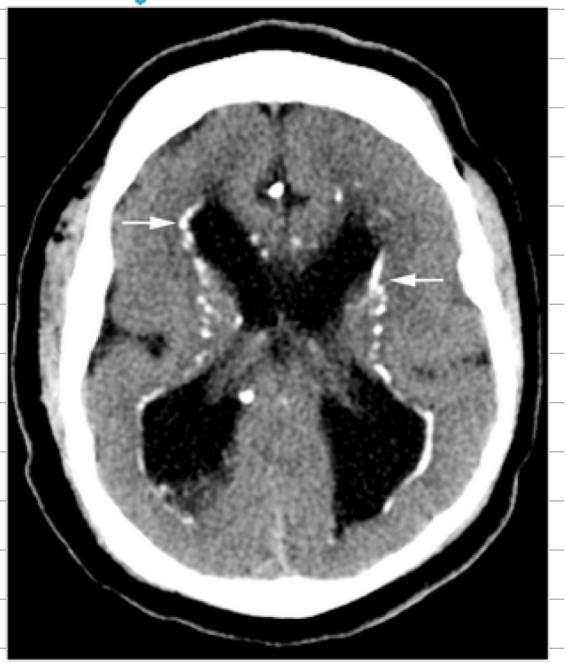
Q. Rx: Ganciclovir (only if baby has progressive neurological disease or SNHL)

↓

I/V ganciclovir for first 6 weeks → oral ganciclovir 6 months

2. Post natal transmission:

- more common
- Transmission through breast milk.
- Baby usually asymptomatic
- infected babies are reservoirs.

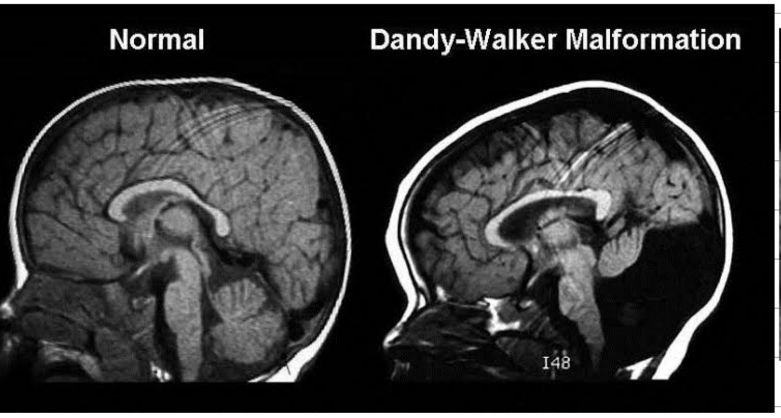


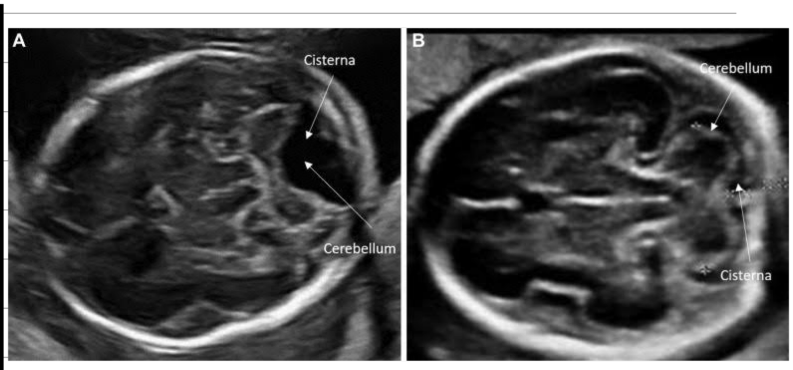
Dandy Walker malformation :

- Defect in the development of the roof of the 4th Ventricle
- This Leads to Expansion of the posterior fossa.

Features :
- The Prominence of occiput is seen (bulding)
- Transillumination is positive.
- Cerebellar Hypoplasia : Ataxia, dysarthria, nystagmus, Vertigo.

Brain imaging : The characteristic fluid-Filled Lesion is the posterior fossa of the brain.
The size of the ventricles is increased.



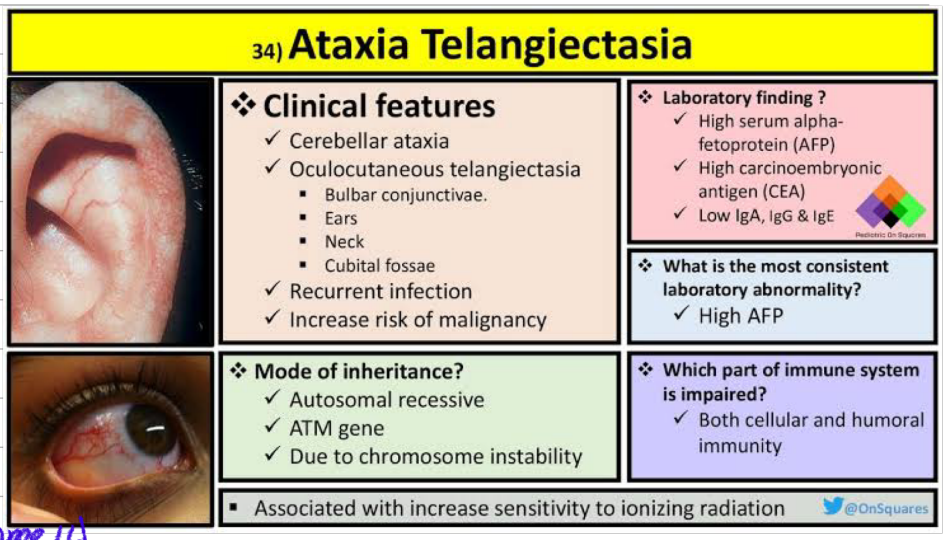


Ataxia Telangiectasia :

- Autosomal recessive disorder.
- Incidence : male = female

Pathogenesis : Defect in ATM gene on Chromosome 11

WT gene for wilm's Tx is also present on chromosome 11.

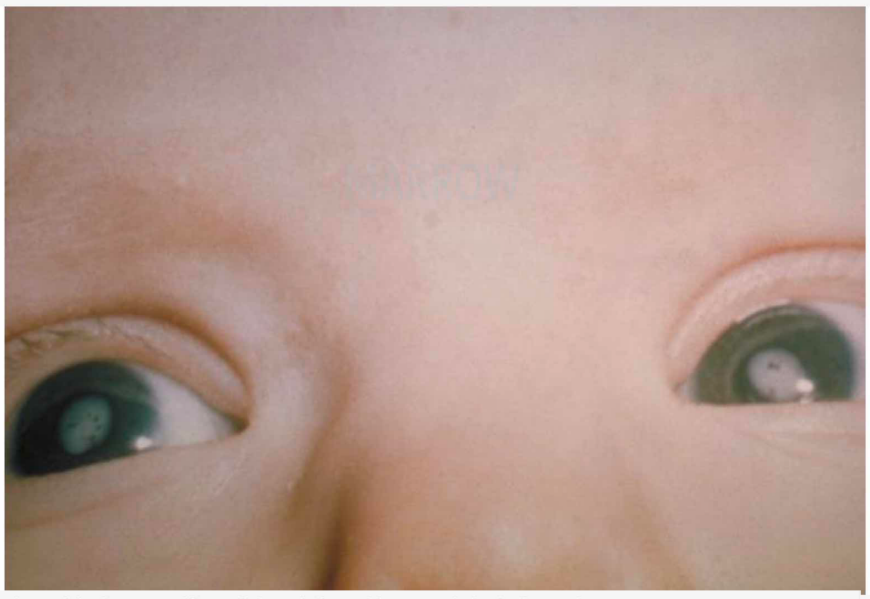


- Normal ATM gene acts as DNA Repair Sensor and activates P53 if any DNA damage occurs.
- Defective ATM gene does not activate P53 and may cause:
  - Ataxia telangiectasia
  - Malignancy
  - Premature aging
  - Neurodegenerative disorders.

Congenital rubella syndrome:

- The triad of Gregg in congenital rubella syndrome includes:
  - **Sensorineural hearing loss -** Nerve deafness is the **single most common** finding among infants with congenital rubella syndrome
  - Congenital **cataract**
  - **Congenital heart disease** - The most common cardiac defect is **patent ductus arteriosus**
- **Maximum damage** to the fetus occurs in the **first trimester**
- Infection in the **second trimester** causes **only deafness**
- If infection occurs **after 16 weeks** there are **no major abnormalities**
- Diagnosis - Presence of IgM rubella antibodies in the infant shortly after birth or persistence of IgG antibodies for >6 months.

The given image shows **bilateral cataract** in an infant with congenital rubella syndrome



The given image shows **blueberry muffin rash** in an infant with congenital rubella syndrome

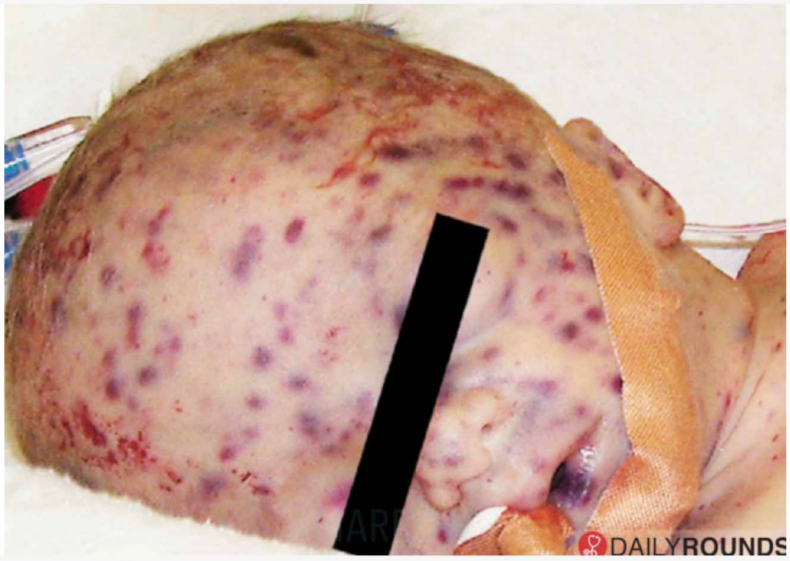
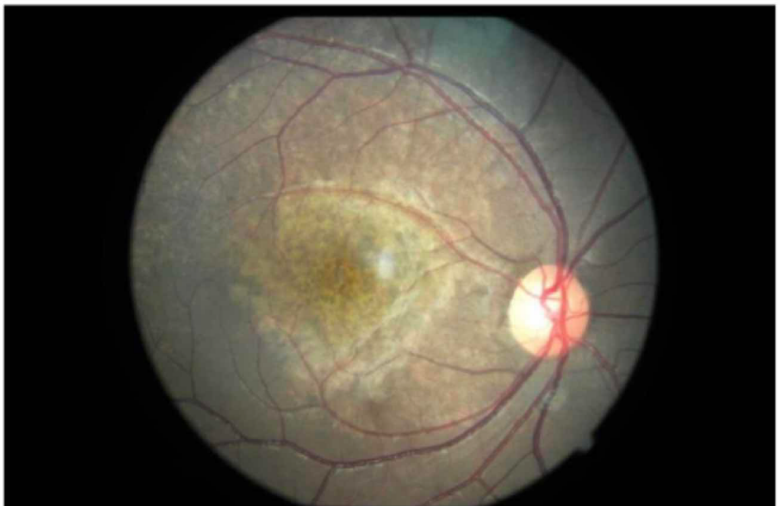


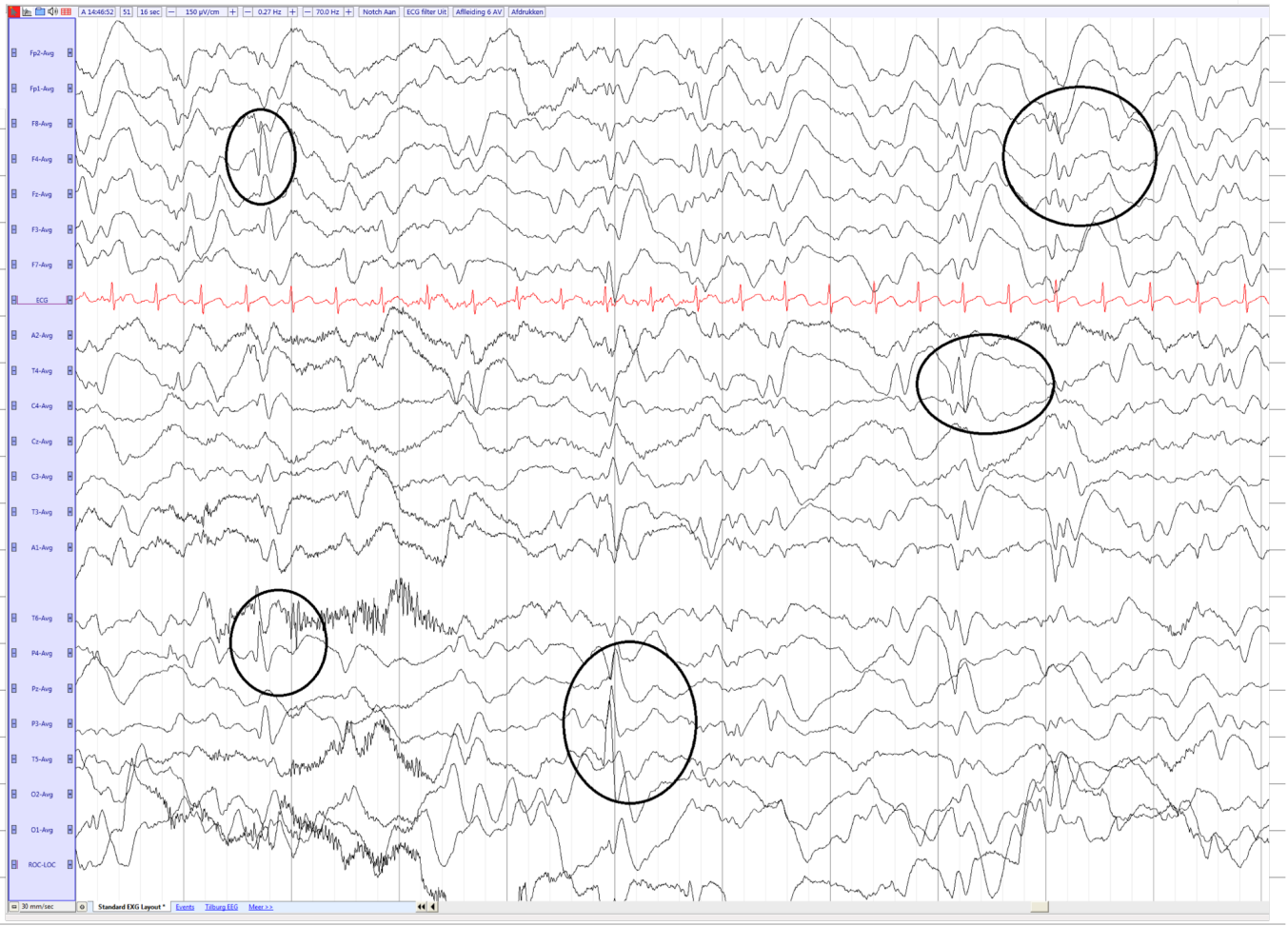
Image Attribution and License

The image given below shows the **salt and pepper fundus.**



West syndrome

- Starts between the ages of 2 and 12 months
- **Triad** of infantile spasms that usually occur in clusters (particularly in drowsiness or upon arousal), developmental regression, and a typical EEG picture – hypsarrhythmia.
- Either cryptogenic or symptomatic
- *ARX* gene (ambiguous genitalia)
- Medical emergency (critical period of 3 weeks)

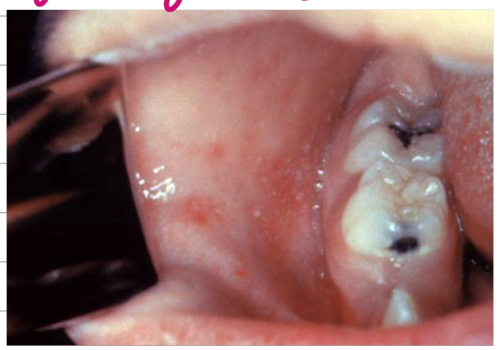


Measles

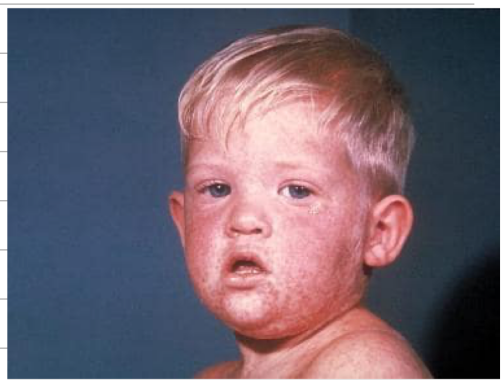
- viral infection : Single-stranded RNA virus of family.
  - Parvoviridae.
  - Incubation period — 8-12 days
  - Secondary attack rate — >90%, High infectivity
  - Infectivity period — 4 days before the appearance of rash to 5 days after the appearance of rash.
- Characteristic features → initially by day 1-3 fever and prodromal features (cough, conyza, conjunctivitis)
- Complications :
  - m/c acute complication — otitis Media
  - most severe complication — Bronchopneumonia.

Day 1-3 fever and prodromal features
↓
Day 2-3 koplik spots
↓
Day 4 Rash (next 4days)
↓
On day 8 Rash disappear
↓
Complications

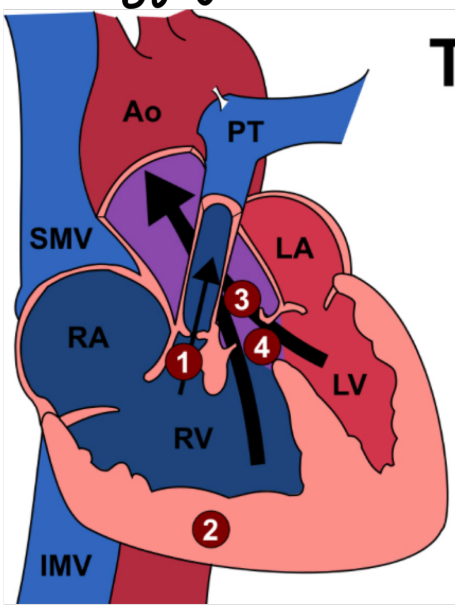
Koplik's spots →

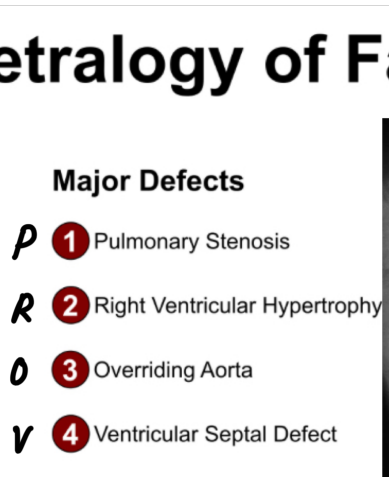


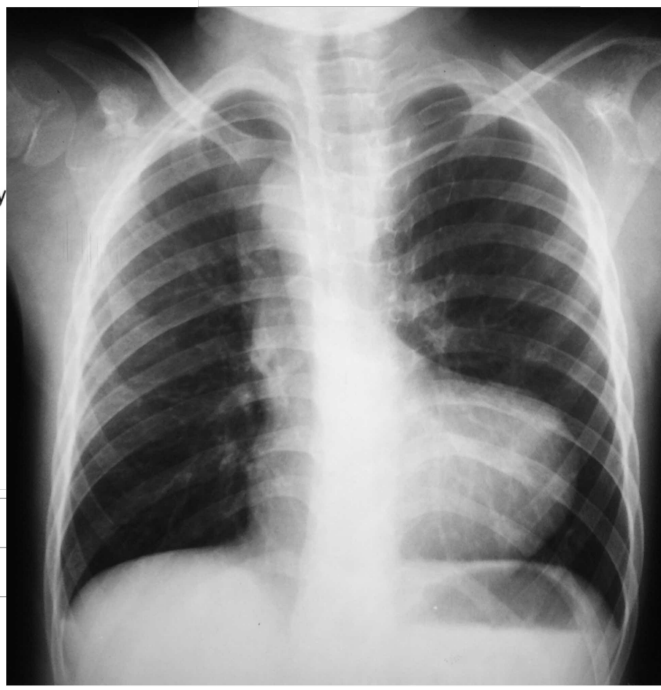
Maculopapular rash →



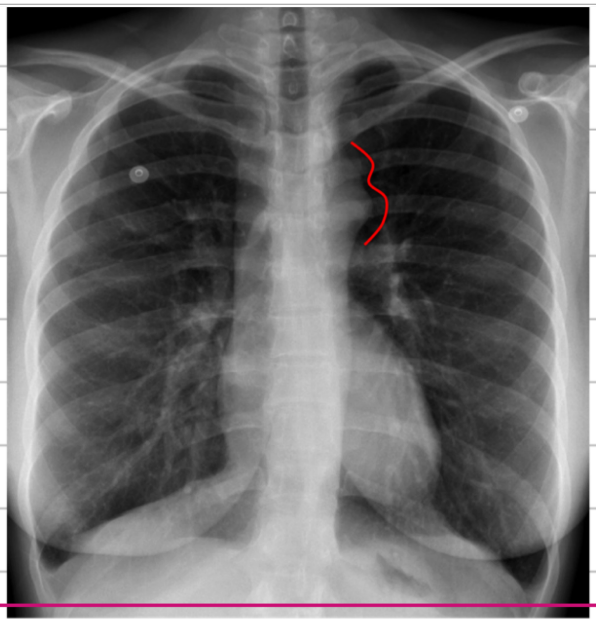
Tetrology of Fallot



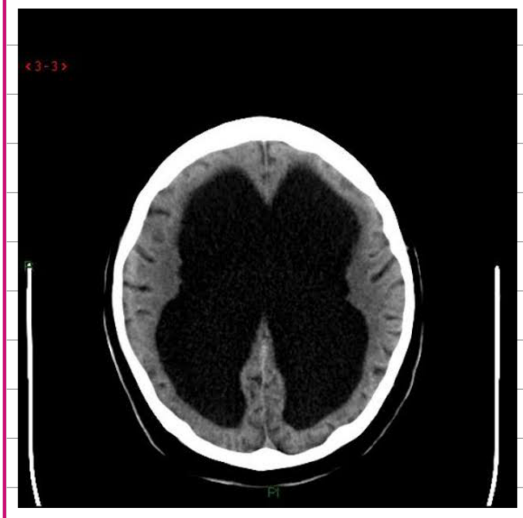


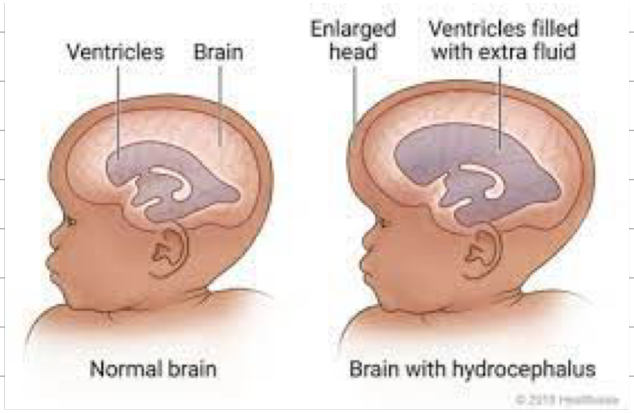


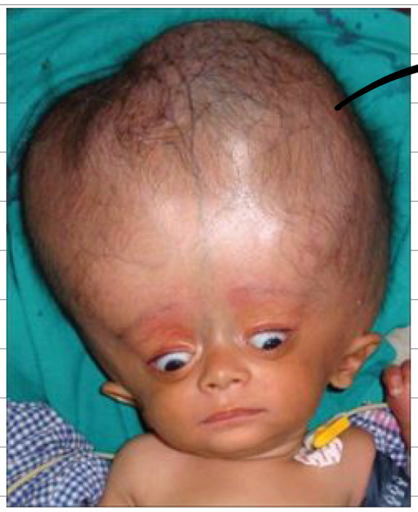
Coarctation of aorta



Congenital Hydrocephalus





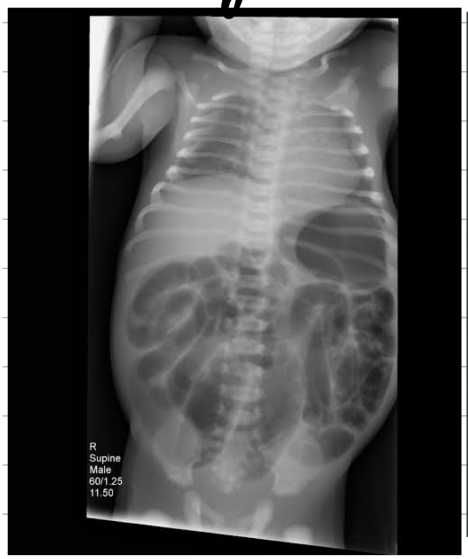


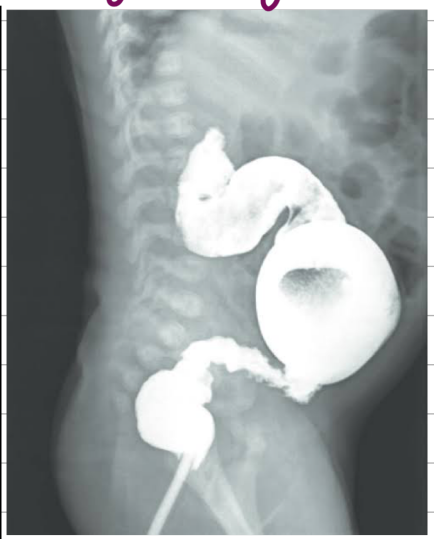
Sunset sign : Eyeball is rotated downwards and the upper part of the sclera is visible.

-Dilated suprapineal recess causes compression on the tectum of the midbrain.

Hirschsprung disease

aka : Congenital megacolon





more common in → Male (4:1, M:F)
most commonly affects
↳ Rectosigmoid area.

IOC : Rectal manometry
⇓
Absence of recto anal inhibitory reflex.

- aka : aganglionosis. → Failure of Relaxation of intestine
- occurs due to failure of migration of neurons during development and is known as
⇓
neurocristopathy.

Alkaptonuria

Homogentisate → Benzoquinone
Homogentisate oxidase ↓ ✗
Maleyl acetic acid (MAA)
Benzoquinone ↓ Polymerise
Alkaptone Bodies

- Enzyme Defect : Homogentisate Oxidase
- accumulate in nose, pinna and sclera : Black spots.

⇒ Homogentisate Excreted in urine, gets oxidised and results in blackish discoloration.
- No mental Retardation.

C/F : Back pain, black spots / Pigmentation in middle age.

Ochronosis : Alkaptone bodies accumulate in intervertebral discs.

Lab diagnosis :
- Alkalinisation of urine : ↑ darkening of urine.
- Ferric chloride test : Positive
- $AgNO_3$ test : Positive
- X-ray Spine : Parrot beak appearance

Rx : Nitsinone / NTBC : inhibits PHPP Hydroxylase and ↓ Homogentisate.

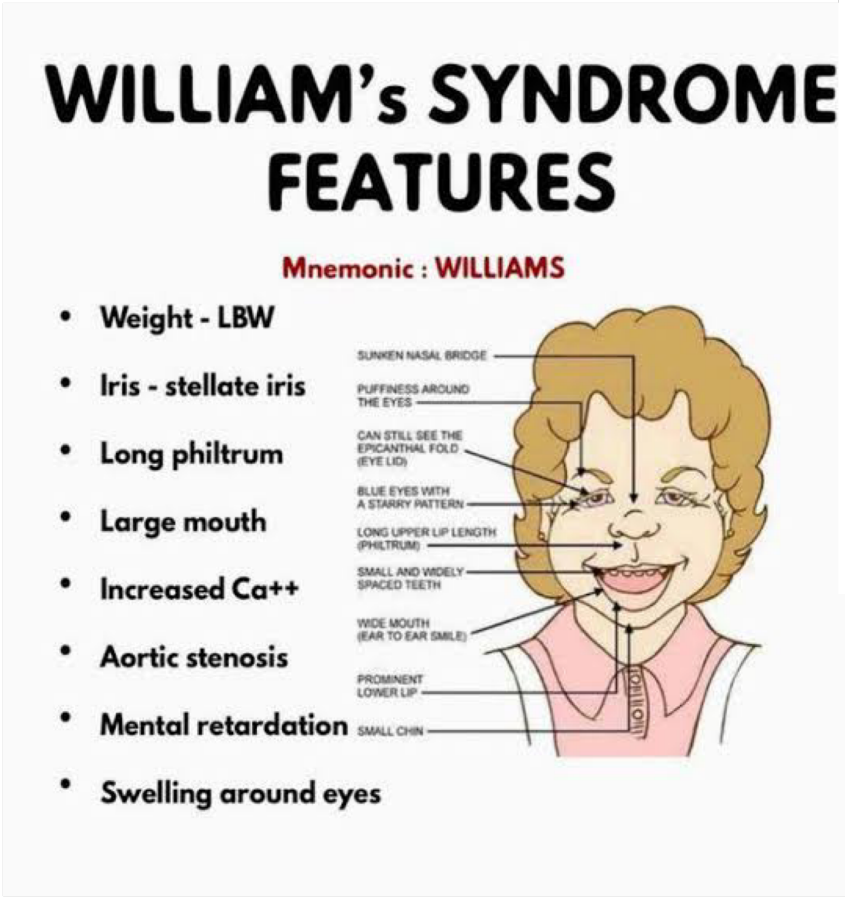
Garrod's Tetrad :
first inborn error of Metabolism studied by Archibald Garrod →
- C - Cysteinuria
- A - Alkaptonuria
- A - Albinism
- P - Pentosuria

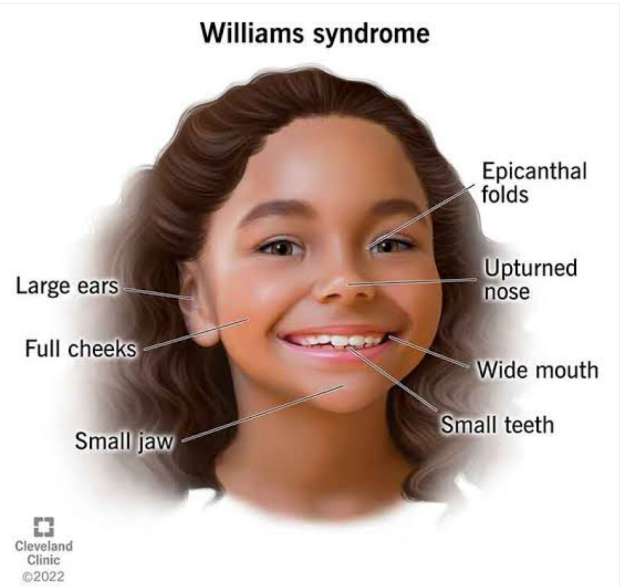
William Syndrome : Microdeletion in 7q 11.23

Facial appearance : • Wide Mouth
• Prominent forehead
• Puffiness in eyes and cheeks.

• These features resemble elves : Elfin facies.
• These children are Happy and Friendly personalities.

Associated Problems : • Hypercalcemia
• Cardiac defect : Supravascular aortic Stenosis.
• Low IQ, Learning disabilities.





Ductus Arteriosus : • Not needed after birth.
• closes within 48-72 hrs (2-3 days)

in some infants, the opening doesn't close. When it stays open → called as Patent ductus arteriousus.

⇓

associated infection : Rubella

Gastroschisis:
- Defect adjacent to umblicus.
- Bowel not covered with peritoneal sac.
- Bowel exposed.
- Less congenital anomalies.
- Atresia and perforation following inflammation of bowel is common.

Omphalocele:
- Defect through the umbilicus in which bowel fails to return inside during embryogenesis.
- Covered with peritoneal sac.
- Large defect, Liver can also Herniate.
- Associated with Beckwith Weidmann syndrome (wilms tumor), Trisomy 13, 18, 21.

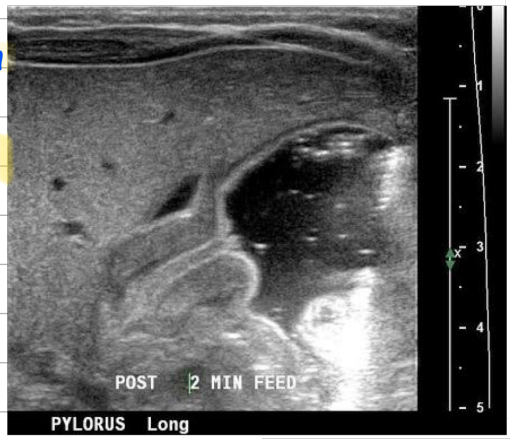
Characteristic	Gastroschisis	Omphalocele
Defect size (diameter)	2-3 cm	2-15 cm
Sac	Never	Always, may be ruptured
Gestational age	Prematurity	Term
Umbilical cord	Adjacent to the defect (defect is seen usually on its right side)	Attached to sac
Herniated viscera	Small bowel, stomach, colon	Small bowel, stomach, colon, liver
Malrotation	Yes	Yes
Bowel character	Inflammatory, edematous	Normal
Enternal nutrition	Delayed	Normal
Associated abnormalities	Uncommon (10% atresia)	Common (50%)

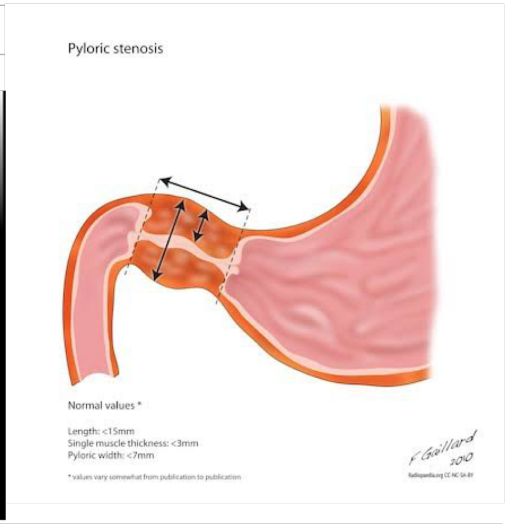
Congenital Hypertrophic Pyloric Stenosis

↓

Lead to gastric outlet obstruction

- M > F
- more common in the first born male child of the family.
- Due to lack of nitrogen oxide Synthase.
- Erythromycine intake early in the life of a child.





HYPERTROPHIC PYLORIC STENOSIS IN A 2 YEARS OLD BOY. Plain radiograph shows severe distension of the stomach with air-fluid level inside due to complete outlet stop. Lateral view on barium exam shows narrowing and elongation of pyloric canal as well as stretching of the duodenal bulb due to hypertrophied pylorus. Notice as ell, the existence of the "string sign" or the "double track sign".

Associations:

- Trisomy 18
- Apert Syndrome
- Cornelia de Lange Syndrome.

C/F : The child is normal at birth.
Symptoms start 2-3wks later.
Projectile non-bilious vomiting.

D/D : Duodenal atresia (Cits present since birth with bilious vomiting)

O/E :- The best time to examine the child is during feeding.

- Visible Peristalsis from left to right
- Followed by Projectile vomiting
- Palpable Olive shaped lump in epigastrium.

on contrast study:

- String Sign
- Double track sign
- Mushroom sign

IOC - USG

↓

Diagnostic criteria:- Thickness more than 4mm

- The Length of the pyloric channel more than 16mm.
- Antral nipple sign.

X-ray → Single bubble sign.

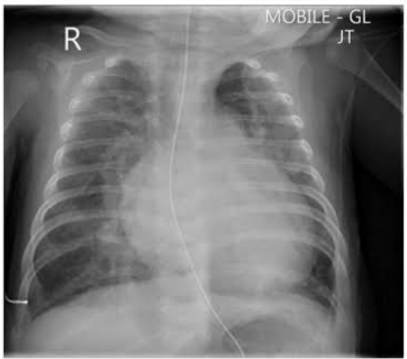
SIGNS	CLASSIFY AS	IDENTIFY TREATMENT (Urgent pre-referral treatments are in bold print)
Two of the following signs: • Lethargic or unconscious • Sunken eyes • Not able to drink or drinking poorly • Skin pinch goes back very slowly	**SEVERE DEHYDRATION**	► If child has no other severe classification - Give fluid for severe dehydration (Plan C). OR If child also has another severe classification: Refer URGENTLY to hospital with mother giving frequent sips of ORS on the way. Advise the mother to continue breastfeeding. ► If child is 2 years or older, and there is cholera in your area, give antibiotic for cholera.
Two of the following signs: • Restless, irritable • Sunken eyes • Drinks eagerly, thirsty • Skin pinch goes back slowly.	**SOME DEHYDRATION**	► Give fluid, Zinc supplements and food for some dehydration (Plan B) ► ***If Child also has a severe classification:*** ***Refer URGENTLY to hospital with mother giving frequent sips of ORS on the way. Advise the mother to continue breastfeeding.*** ► Advise mother when to return immediately. ► Follow-up in 5 days if not improving. ► If confirmed/symptomatic HIV, follow-up in 2 days if not improving.
• Not enough signs to classify as some or severe dehydration	**NO DEHYDRATION**	► Give fluid, Zinc supplements and food to treat diarrhoea at home (Plan A) ► Advise mother when to return immediately. ► *Follow-up in 5 days if not improving.* ► If confirmed/symptomatic HIV, follow-up in 2 days if not improving.

Pulmonary Plethora : is a term used to describe the appearances of increased pulmonary perfusion on chest radiographs.

Pulmonary Plethora

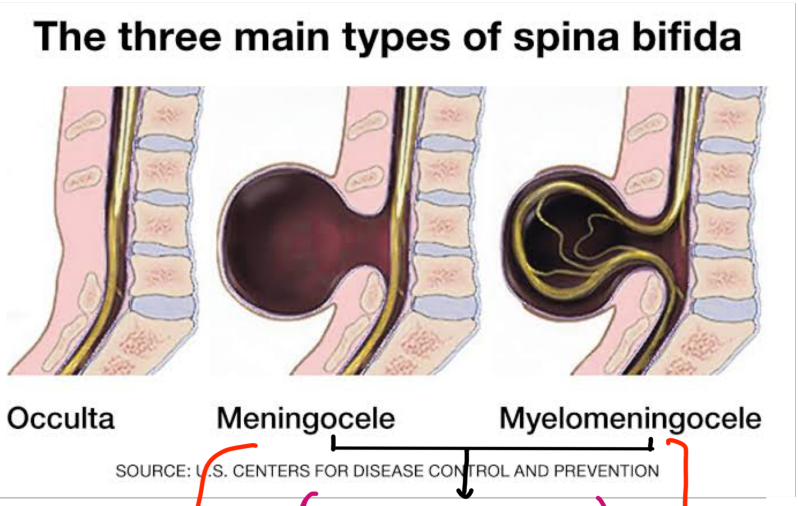
Plain radiograph

- prominent pulmonary vasculature
  - pulmonary vessels are dilated and tortuous extending farther into the peripheral one-thirds of the lungs
  - diameter of a pulmonary artery is greater than the accompanying bronchus
- increased size of number of hilar pulmonary arteries
  - >3-5 end-on should be seen
  - diameter of the right descending pulmonary artery is bigger than the diameter of the trachea
- cardiomegaly may be present



Spina Bifida Occulta

- AKA: close spina Bifida.
- There's a defect in one or more vertebrae.
- There's no Herniation of meninges.
- The site is marked by a tuft of hair.
- Asymptomatic.
- Diagnosis → X-ray



(Spina bifid Aperta)

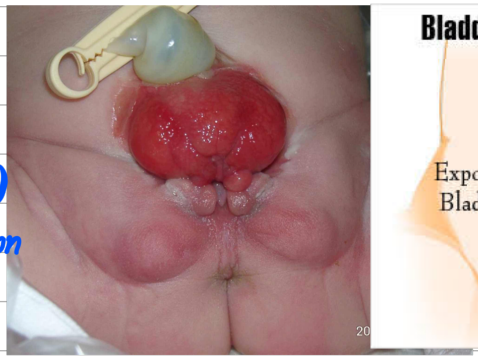
- Protrusion of meninges
- Protrusion and opened spinal cord

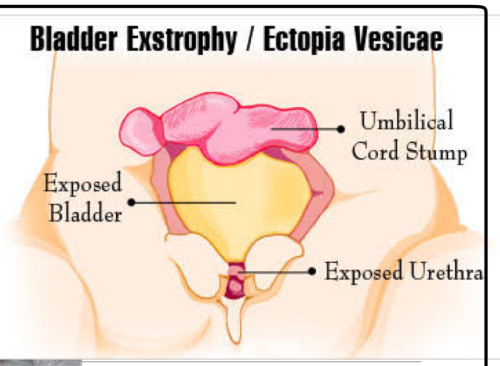
Bladder Extrophy

aka → Ectopia Vesicae

- most severe type of epispadias.
- Pubic diastasis (pubic bones are far apart) iliac osteotomy can be done for correction
- males: undescended testis
- females: Bifid Clitoris

- Bad prognosis.





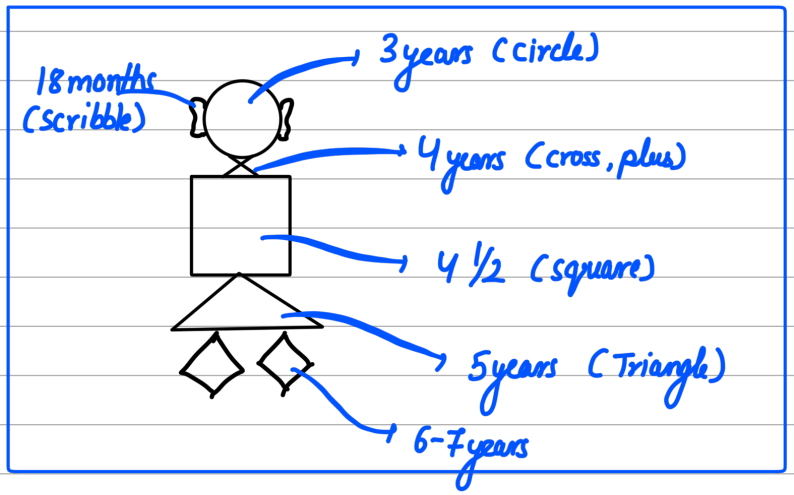
Milestones

(A) Gross Motor Milestone:

- Head control - 1st sign of head control - 6 wks
  - Partial Head control - 3 months (neck Holding)
  - Complete Head control - 5 months → Rolls over → 5 months
- Sitting control - Sits with support (Tripod) - 6 months
  - Sits without support - 8 months
- Stand and walking - Stand with support - 9 months
  - Stand without support - 12 months → Stands without support
  - walk with support - 12 months → Creeps well / walks but walls
  - walk without support - 15 months → Creeps upstairs
- Climb stairs - 2 years (2 feet / step)
- Upstairs on alternate foot (Rides tricycle) 3 years
- Walks downstairs with alternate foot (Hopping on 1 foot) → 4 years
- Skip on both feet - 5 years

(B) Fine Motor Milestones :

- Bidextrous grasp (with 2 hands) - 4 months
- Unidextrous grasp (with 1 hand) - 6 months
- Ulnar grasp - 7 months
- Radial grasp - 8 months
- Pincer grasp ← Immature → 9 months
  ← Mature → 12 months
- Tower of 2 cubes - 15 months
  6 cubes - 2 years
  20 cubes - 3 years
- Scribbling (〰) - 15 months
- Lines (—) - 2 years
- Circle - 3 years
- Cross / plus - 4 years
- Square - 4½ years
- Triangle - 5 years
- Diamond / Rhomboid - 6-7 years

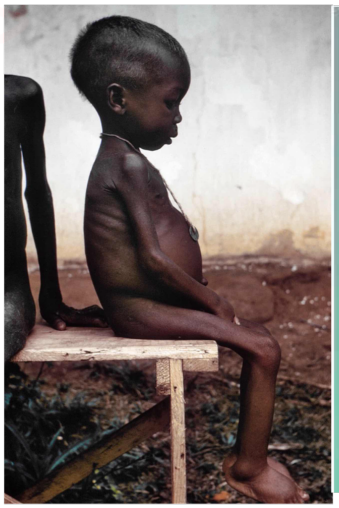


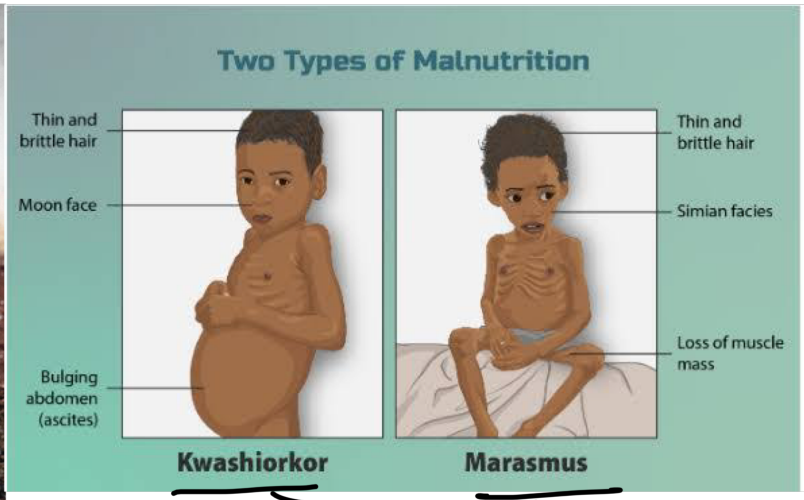
(C) Language milestone :

- Alerts to sound - 1 month
- Cooing - 2 months
- Laughs aloud - 4 months
- Monosyllables (ma, ba) - 6 months
- Bisyllables (mama, baba) - 9 months
- 1-2 words 1 meaning - 12 months
- 8-10 word vocabulary - 18 months
- 2-3 word sentences - 2 years
  (use pronouns like I, me, you)
- Asks Questions and knows name and gender - 3 years
- Sings songs, poem and tells stories - 4 years
- Asks meaning of words - 5 years

(D) Social milestones :

- Social smile - 2 months
- Recognizes mother - 3 months
- Stranger anxiety - Stranger anxiety
- Waves bye-bye - Waves bye-bye
- Comes when called, Plays simple ball game - 12 months
- Jargon - 15 months
- Copies parents in task - 18 months
- Asks for food, drink, toilet - 2 years
- Shares toys, Knows full name and gender - 3 years
- Plays cooperatively in a group and goes to toilet alone - 4 years
- Helps in household task and can dress and undress on its own - 5 years





Differences between Marasmus and Kwashiorkar

Marasmus (more common)	Kwashiorkor
Seen in infants less than 1 year of age	Seen in older children (6 months to 3 years of age)
Total calorie deficit	Protein deficit
No edema	Edema is prominent
Voracious appetite No apathy	Loss of appetite Apathy
Severe muscle wasting	Muscle wasting mild or absent
Prominent ribs	Protuberant abdomen
No Fatty liver No subcutaneous fat	Fatty liver Subcutaneous fat preserved
Special features: 1. Alert 2. Loose wrinkled skin 3. Baggy pant appearance	Special features: 1. Moon face 2. Flaky paint dermatosis 3. Flag sign

Malnutrition

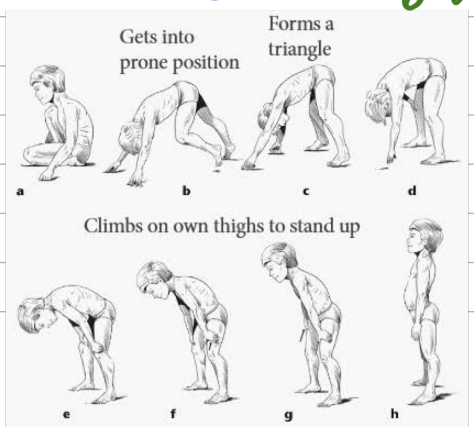
- **Duration**
  - **Acute** → Weight is reduced: **Wasting** → Weight-for-height chart (W/H)
  - **Chronic** → Height is affected: **Stunting** → Height-for-age chart (H/A)
- **Based on severity**
  - **Moderate** → −2SD to −3SD (standard deviation) → B/L pedal edema → (NO)
  - **Severe** → <−3SD → (Yes)

Duschene Muscular Dystrophy

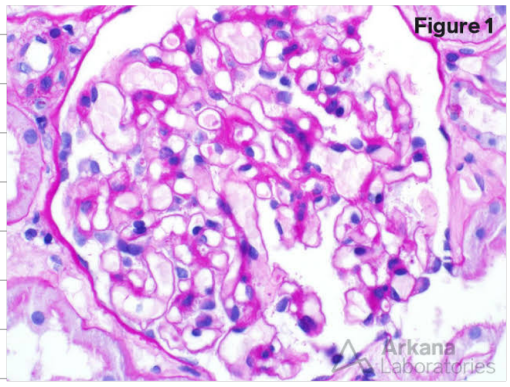
- X-linked recessive (female carriers, only males affected)
- Defect in short arm of chromosome Xp21
- Dystrophin protein defective → Dystrophinopathies
  Dystrophin → for coordination of muscular contraction.
- Based on severity of defect: • Frameshift / Nonsense (major) → DMD
  • Miss Sense (minor) → Becker's muscular dystrophy

- Features: - Onset < 5 years
  - Noticed when child starts walking or running around two years of age.
  - Delay in attaing motor milestones (walk, run).
  - Characterised by Proximal muscle weakness.
  - Muscles affected → Thigh, calf (waddling gait)

Gower sign → Positive

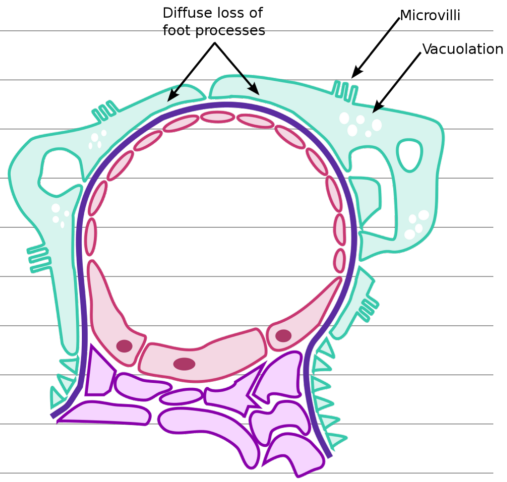


Minimal Change disease



- aka Lipoid nephrosis.
- 90% of nephrotic Syndrome in children are MCD.
- Peak in children: 2-7 (Boys > Girls)

Presentation :
- Edema
- Selective proteinuria
- Hyperlipidemia
- Lipiduria
- Hypoalbuminemia.



AKA called as 100% as the patient will present itself in one way only, no other presentation.

Rx: oral Prednisolone (2mg/kg/day)

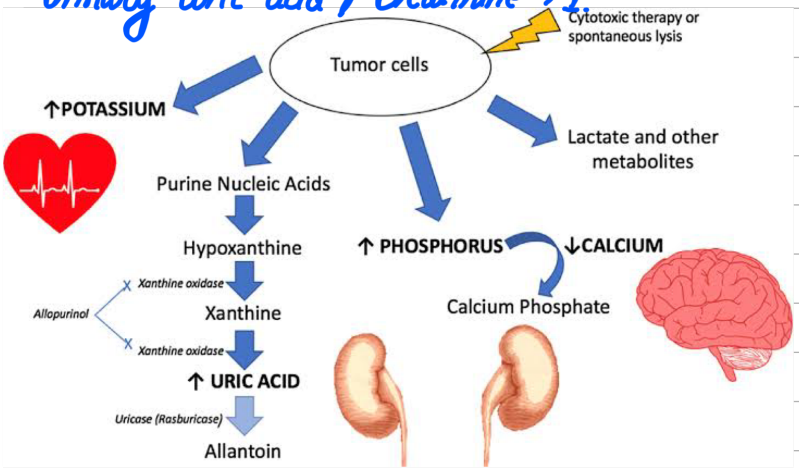
Tumor lysis Syndrome

- It is an oncological emergency.
- It is also called Uric acid nephropathy.

Tumor lysis syndrome is a clinical condition that can occur spontaneously or after the initiation of chemotherapy associated with the following metabolic disorders: hyperkalemia, hyperphosphatemia, hypocalcemia, and hyperuricemia leading to end-organ damage. It is most common in patients with solid tumors

Rx: Hemodialysis

- Uric acid will precipitate inside tubular Lumen → Oliguric renal failure → Tumour Lysis Syndrome.
- Urinary uric acid / Creatinine >1



Nephrotic syndrome

It is a combination of:

1. Nephrotic Proteinuria
2. Hypoalbuminemia
3. Edema
4. Hyperlipidemia
5. Lipiduria

- Minimal change disease is a podocytopathy (injury to podocyte) leading to Proteinuria.
- Diabetes Mellitus is the Most common cause of Nephrotic range of Proteinuria.

Presentation in children:

- Severe edema (Pedal edema and facial puffiness)
- Urine Albumin : 3+
- S. Albumin : Low
- Total Cholesterol : Very high