Pediatrics Mini-OSCE

2023 Edition

إعداد محمود بركات


But ....But..
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أصغر من دوسية الجلدية

الملاحظات

- شامل لأسئلة سنوات حتى 27/12/2022
- الملف مقسم حسب المحاضرات والمحاضرات مقسمة حسب السستمات وقبل كل موضوع تم الإشارة الي درجة تكرار الموضوع كتالي:
  - "High-Yield" وهي المواضيع الي بتيجي بمعظم الامتحانات
  - "Medium-Yield" وهي المواضيع الي بتيجي بشكل متكرر
  - "Low-Yield" وهي المواضيع الي بتيجي بشكل غير متكرر
- أسئلة السنوات المكررة تم جمعها بسؤال واحد ووضع عدد مرات تكرار السؤال في هامش أعلى الصفحة من جهة اليمين
- أي كتابة بصندوق أزرق يعتبر هامش للملاحظات وأي كلام بالأزرق يعد إضافي من عندي

Last edit: 21/Oct/2023

Growth & Development

Developmental Assessment

High-Yield

Developmental Assessment

❖How to adjust for prematurity ?

- **During the first year**: Corrected age = Chronological age - (9 - # of months he spend in the womb)
- **During the second year**: Corrected age = Chronological age - ½ (9 - # of months he spend in the womb)
- Correction is not required after about 3 years of age

❖What are the 4 areas of development ?

1. Gross motor
2. Fine motor & vision
3. Hearing, Speech, & Language
4. Behavioral, Emotional, & Social

Milestones

Age	Gross motor		Fine motor & vision	Hearing & speech	Social	Vaccine
	Prone	Supine				
< 1 month	Symmetrical flexion of limbs	Marked head lag	1. Follows with eyes to medline only 2. Hands tightly fisted	Startles to loud noise	Newborn: fix on face Spontaneous smile	BCG
2 months **Vac at 61**	Raise head to 45°	Start holding head	1. Move object or face 180° 2. Briefly retains rattles	–	Responsive smile Recognize parents	1. Hexa – 1st 2. Rota – 1st
3 months **Vac at 91**	Raise half chest	No head lag	–	Vocalizes, laughs and coo	–	1. Hexa – 2nd 2. Rota – 2nd 3. OPV – 1st
4 months **Vac at 121**	Raise whole chest (4-6 months)	Pulls to set (3-6 months)	1. Reaches out for toys with both hands 2. Bats at objects 3. Grabs and retains objects	Orients to voice, laughs and squeals	Initiates social interaction	1. Hexa – 3rd 2. Rota – 3rd 3. OPV – 2nd

شرح

Milestones

Age	Gross motor	Fine motor & vision	Hearing & speech	Social	Vaccine
6 months	1. Raise whole chest 2. Sits momentarily with round back 3. Rolls in both directions	1. Reaches with one hand 2. Transfers toys from one hand to the other 3. Palmar grasp	1. Babbles ‘da’, ‘ma’ 2. Hearing distraction test	Recognize object or person as unfamiliar	–
8 months	Sits with straight back	–	–	Puts food in mouth (بحط كل شيء بفمه)	–
9 months Vac at 271	1. Crawling (8-9) 2. pulls to stand	Object permanence	1. Dada, Mama 2. Understand No	1. Plays gesture games 2. Understands own name 3. Stranger anxiety	1. Measles 2. OPV – 3rd 3. Vit A 100 thousand
10 months	1. Stands with support 2. Walks around furniture	Mature pincer grasp	-	1. Waves bye-bye 2. Plays “peek-a-boo”	–

Milestones

Age	Gross motor	Fine motor & vision	Hearing & speech	Social	Vaccine
12 months	Walks unsteadily, broad gait, hands apart	1. Casting the objects 2. Can voluntarily release items	1. 2-3 words other than dada/mama 2. Commands with gesture	1. Drinks from a cup 2. Comes when called 3. Cooperate with dressing	1. MMR-1 2. HAV
15 months	Walks alone steadily	1. Scribbles with pencils (14-15 months) 2. Makes tower of 2 cubes	1. 4 to 6 words in addition to above 2. uses jargon 3. respond to one step verbal demand	1. indicates desires by pointing	–
18 months	1. Runs, walks up stairs with handheld, 2. Stoops and recovers	1. Uses spoon 2. Makes tower of 3 cubes	1. 10 single words 2. shows 4 parts of the body **21 months**: use two or more words to make simple phrases	1. holds spoon, 2. symbolic play, 3. kisses parents, 4. tells mother she/he wants potty 5. Plays near (but not with) other children	1. MMR-2 2. DaPT 3. OPV 4. HAV 5. Vit A 200 thousand

Milestones

Age	Gross motor	Fine motor & vision	Hearing & speech	Social
2 years	1. Runs, walks, climbs stairs (2 feet/step) 2. Kicks ball, throw ball overhand 3. Jumps with 2 feet off the floor	1. Uses fork 2. Copies a straight line 3. Makes tower of 6 cubes	1. Vocabulary 50 words 2. 2-to-3-word phrases 3. Uses I and me 4. 50% of speech intelligible to stranger	1. Dry by the day 2. Pulls off some clothing 3. Listens to stories with pictures **30 months**: full name
3 years	1. Climbs stairs (1 foot/step) 2. Pedal tricycle, broad jumps	1. Copies a circle 2. Makes a bridge with cubes	1. Speech in sentences 2. Vocabulary 200 words 3. Uses 5-to-8-word sentences 4. 75% of speech intelligible to stranger	1. Interactive play 2. Takes turns 3. Knows age and gender
4 years	1. Goes up- & down-stairs (1 foot/step), run upstairs 2. Balances on one foot	1. Copies a cross 2. Catches ball 3. Makes steps with cubes	1. Count to 20 or more 2. Tells a story 3. 100% of speech intelligible to stranger	1. Goes to the toilet alone 2. Brush teeth 3. Washes and dries hands 4. Imaginary play

شرح

Milestones

Age	Gross motor	Fine motor & vision	Hearing & speech	Social
5 years	Skips with alternating feet	1. Draws a person with 6 body parts 2. Copies a square	1. Grammatical speech 2. Asks what words mean	1. Dresses & undresses alone 2. Name 4 colors 3. Plays cooperative games 4. Understands rules and abides by them
6 years	Rides a bike	Writes name	Identifies written letters and numbers	1. Knows right from left 2. Knows all color names

ملاحظات

- لكل امتحان ميني اوسكي تقريبا 4 اسئلة على هذا الموضوع
- السؤال بكون يا على أعمدة الجدول السابق بمعنى تقارن بين التطور في مجال معين أو بكون على الأسطر بمعنى يسألك عن مستوى تطور الطفل في باقي المجالات مثلا بقلك الطفل بقدر يقعد لحاله وبسألك شو بقدر يسوي في مجال السمع مثلا
- بعض الأسئلة مركبة بين المحاضرات مثلا بقلك هذا الطفل الي بياكل بملعقة شو المطاعيم الي لازم ياخذها

Milestone – Newborn

- **What is the developmental age ?**
  - Less than 6 weeks
- **What is the name of this skill ?**
  - Marked head lag
- **When dose it disappear ?**
  - Around 3 months
- **What skill would this baby show on prone position ?**
  - Limbs flexed symmetrically

Milestone – Newborn

- **What is the age ?**
  - Less than 6 weeks
- **What is the name of this skill ?**
  - Spontaneous smile
- **What does hearing assessment of this baby show ?**
  - Startle to loud noises

Milestone – 6 weeks

- **What is the age ?**
  - More than 6 weeks
- **What is the name of this skill ?**
  - Responsive smile
- **What other fine motor does this child have ?**
  - follow object in 180 degree
- **What other gross motor does this child have ?**
  - Raise head to 45^{o}
  - Start holding head

Milestone – 3 months

- **What is the age ?**
  - 3 months
- **What is the name of this skill ?**
  - Raises half of the chest

- **Masa lefts her head, says goo, ahh**
- **What is the age ?**
  - 3 months
- **What is the name of this skill ?**
  - Vocalizes and coo

Milestone – 3 months

- **How old is the baby ?**
  - 3 months
- **How many words can he speak ?**
  - Vocalizes, laughs and coo
- **DDx for the rash**
  - Chickenpox
  - Measles
  - Rubella
  - HSV

Milestone – 4 months

- **What is the age ?**
  - 4 months
- **What is the name of this skill ?**
  - Reaching out with both hands
- **What skill this baby has on prone position ?**
  - Raises half of his chest
- **What other fine motor does this child have ?**
  - Bats at objects
  - Grabs and retains objects

Milestone – 6 months

- **What is the age ?**
  - 6 months
- **What is the name of this skill ?**
  - Raise all the chest on Palm
- **What skill this baby has on supine position ?**
  - Sits momentarily with round back
- **What fine motor skills does this baby has ?**
  - Reaches with one hand
  - Transfers toys from one hand to the other
  - Palmar grasp

Gross motor

1. Raise whole chest
2. Sits momentarily with round back
3. Rolls in both directions

Milestone – 6 months

- **The age of this baby not more than ?**
  - 6 Months
- **Why ?**
  - Because he sits with rounded back
- **What skill this baby has on prone position ?**
  - Raise all the chest on Palm
- **How to assess hearing in this baby ?**
  - Hearing distraction test

Hearing distraction test

- **What's the name of this test ?**
  - Distraction test
- **What does it test for ?**
  - Auditory function, hearing, cochlear branch of vestibulo-cochlear nerve
- **What is the age ?**
  - 6-18 months

Milestone – 8 months

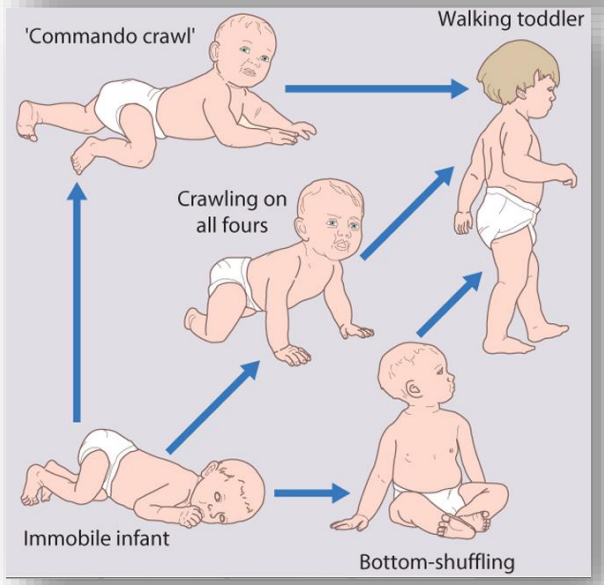
- **What is the age ?**
  - 8 months
- **What is the name of this skill ?**
  - Mouthing
- **What other skill is seen at this age ?**
  - Sit with straight back

Milestone – 8 months

- **What is the age ?**
  - 8 months
- **What is the name of this skill ?**
  - Sit with straight back
- **What other skill is seen at this age ?**
  - Mouthing

Milestone – 8-9 months

- **What is the age ?**
  - 8-9 months
- **What is the name of this skill ?**
  - Crawling



Milestone – 9 months

- **What is the age ?**
  - 9 months
- **What is the name of this skill ?**
  - Object permanence
- **What speech skills does this baby has ?**
  - Dada, Mama
  - Understand No

Milestone – 9 months

- **What is the age ?**
  - 9 months
- **What is the name of this skill ?**
  - Pat a cake
- **What other social skills does this baby has ?**
  - Understands own name
  - Stranger anxiety

Milestone – 9 months

- **What is the age ?**
  - 9 months
- **What is the name of this skill ?**
  - Stranger anxiety
- **What vaccines should this toddler had taken at this age ?**
  - Measles
  - OPV (third dose)
  - Vitamin A (100 thousand international units)

Milestone – 10 months

- **What is the age ?**
  - 10 months
- **What is the name of this skill ?**
  - Mature pincer grasp
- **What gross motor skill is associated with this fine motor skill ?**
  - Walk around with support (furniture)

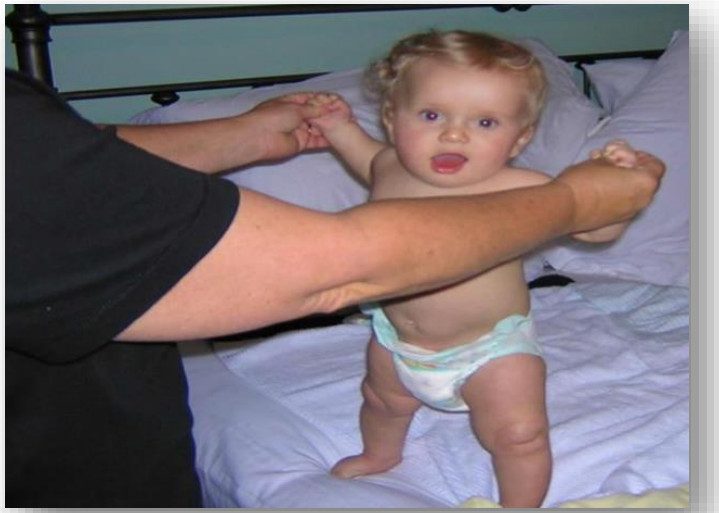
Milestone – 10 months

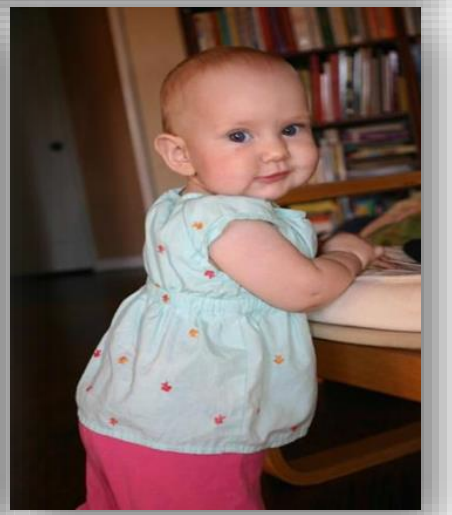
❖**This child will be able to perform all skill except ?**

a. Stand with the support
b. Wave bye-bye
c. Understand own name
d. Cast object
e. Object permanence

Milestone – 10 months

- **What is the age ?**
  - 10 months
- **What is the name of this skill ?**
  - **First picture**: Stands with support
  - **Other pics**: Walk around with support (furniture)
- **What fine motor skill is associated with this gross motor skill ?**
  - Mature pincer grasp





Milestone – 10 months

- **What is the age ?**
  - 10 months
- **What is the name of this skill ?**
  - Waves bye-bye
- **How much words can she say ?**
  - Mama, Dada
- **What type of games can they play ?**
  - Plays gesture games
  - Plays “peek-a-boo”

Milestone – 12 months

- **What is the age ?**
  - 12 months
- **What is the name of this skill ?**
  - Casting the objects
- **Mention another fine motor skill ?**
  - Can voluntarily release items
- **What gross motor skill is associated with this fine motor skill ?**
  - Walks unsteadily, broad gait, hands apart

Milestone – 14-15 months

- **What is the age ?**
  - 14-15 months
- **What is the name of this skill ?**
  - Scribbling with pencil
- **How he tell his mother thing that he wants ?**
  - Pointing to it

Milestone – 18 months

- **What is the age ?**
  - 18 months
- **What is the name of this skill ?**
  - Using spoon
- **No. of cubes can build ?**
  - 3 cubes
- **How he tell his mother thing that he wants ?**
  - Pointing to it
- **How many words can he speak ?**
  - 10 words

Milestone – 18 months

❖**What's the vaccines taken at this developmental age ?**

1. MMR-2 – second dose
2. DTaP – the supportive dose
3. OPV – the supportive dose
4. Vitamin A (200 thousand international units)
5. HAV (second dose)

Milestone – 18 months

- **What is the age ?**
  - 18 months
- **What is the name of this skill ?**
  - Shows 4 parts of the body
- **How many words he can say ?**
  - 10 words
- **Can this kid use spoon or fork ?**
  - He can use a spoon

Milestone – 18 months

- **What is the age ?**
  - 18 months
- **What is the name of this skill ?**
  - Symbolic play
- **How many words he can say ?**
  - 10 words

Milestone – 18 months

- **What is the age ?**
  - 18 months
- **What is the name of this skill ?**
  - Stoops and recovers

Milestone – 2 years

- **What is the age ?**
  - 2 years
- **What is the name of this skill ?**
  - Listens to stories with pictures
- **How many words he can say ?**
  - 50 words

Milestone – 3 years

- **What is the age ?**
  - 3 years
- **What is the name of this skill ?**
  - Pedal tricycle
- **What part of development is assessed in this image ?**
  - Gross motor

Milestone – 3 years

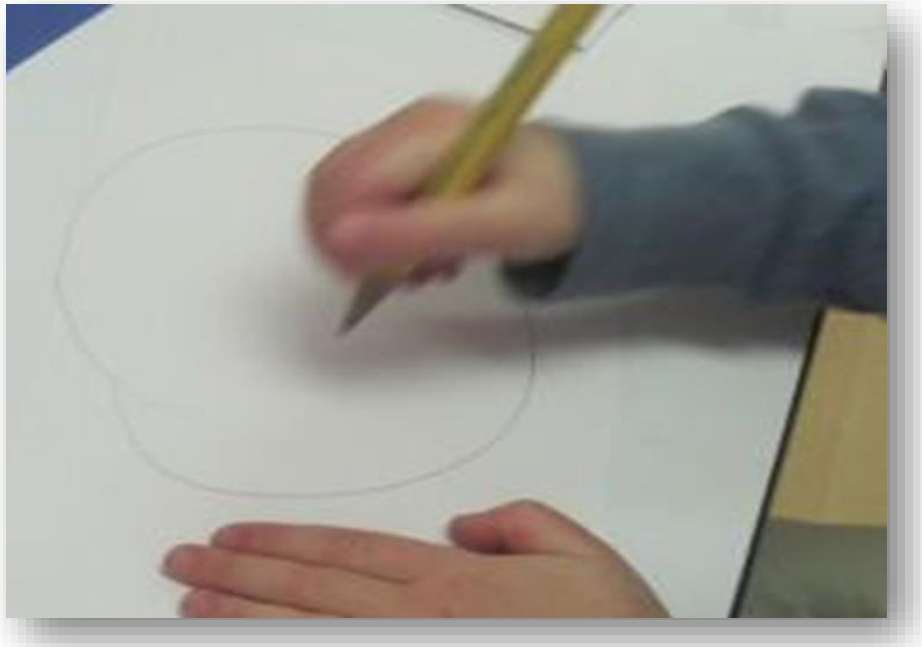
- **What is the age ?**
  - 3 years
- **What is the name of this skill ?**
  - interactive play

Milestone – 3 years

- **What is the age ?**
  - 3 years
- **What is the name of this skill ?**
  - Broad jump

Milestone – 3 years

- **What is the age ?**
  - 3 years
- **What is the name of this skill ?**
  - Draw a circle



Milestone – 4 years

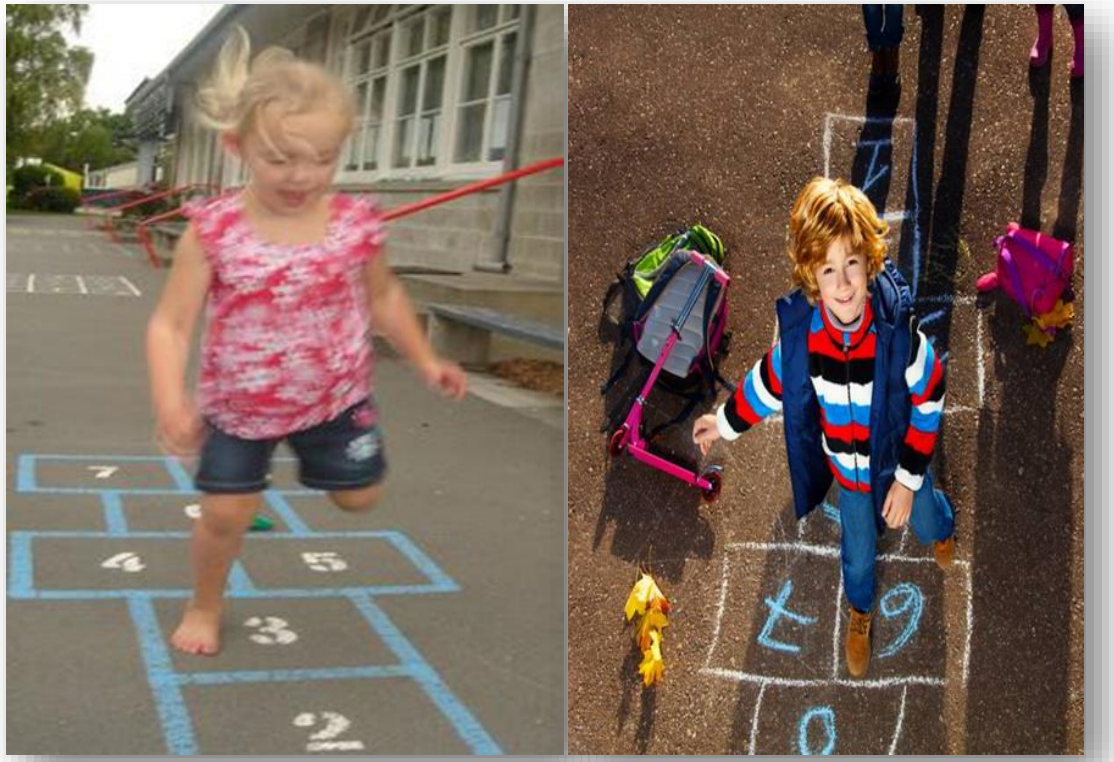
- **What is the age ?**
  - 4 years
- **What is the name of this skill ?**
  - Brush teeth
- **What's her pencil skill ?**
  - Draws a cross
- **How many can she kid count ?**
  - Count to 20
- **How many colors can she name ?**
  - 4 colors

Milestone – 4 years

- **At what age would you expect a child to stand on one foot ?**
  - 4 years

Milestone – 5 years

- **What is the age ?**
  - 5 years
- **What is the name of this skill ?**
  - Skips with alternating feet
- **What's their pencil skill ?**
  - Draws a triangle



Milestone – 5 years

- **At what age would you expect a child to dress himself ?**
  - 5 years

Gross motor – Prone

Age	Gross motor – Prone	
Newborn	Symmetrical flexion of the limbs	
6-8 weeks	Raises head to 45 degree	
3 months	Raises half of the chest	
6 months	Raises the whole chest on palms	

Gross motor – Supine

Age	Sitting	Age	Walking
Newborn	Marked head lag	10 months	1. Stands with support 2. Walks around furniture
6-8 weeks	Start holding head	12 months	Walks unsteadily, broad gait, hands apart
3 months	No head lag	15 months	Walks alone steadily
3-6 months	Pulls to set	18 months	Runs, walks up stairs with handheld
6 months	Sits momentarily with round back	2 years	Runs, walks, climbs stairs (2 feet/step)
8 months	Sits with straight back	3 years	Climbs stairs (1 foot/step)
9 months	1. Crawling (8-9) 2. Pulls to stand	4 years	Goes up- & down-stairs (1 foot/step), run upstairs

Gross motor – Supine

- **At what age you expect a child to walk independently ?** (Give arange)
  - 11 months (25%) – 18 months (97.5%)

- **A boy who says few words other than (mama, dada, baba), has just started to walk and has mature pincer grasp .What is the age of this child?**
  - 1 year old

Gross motor

Age	Jumping & balance	Age	Balls & Bikes
18 months	Stoops and recovers	15 months	Throws ball underhand
2 years	Jumps with 2 feet off the floor	2 years	Kicks ball Throws ball overhand
3 years	Broad jumps	4 years	Catches ball
4 years	Balances on one foot	3 years	Pedal tricycle
5 years	Skips with alternating feet	6 years	Rides a bike

Fine motor – Grasps

Newborn

Palmer reflex (Hands tightly fisted)

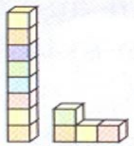
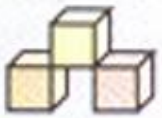
6 months

Palmer grip

10 months

Pincer grip

Fine motor – Building with bricks

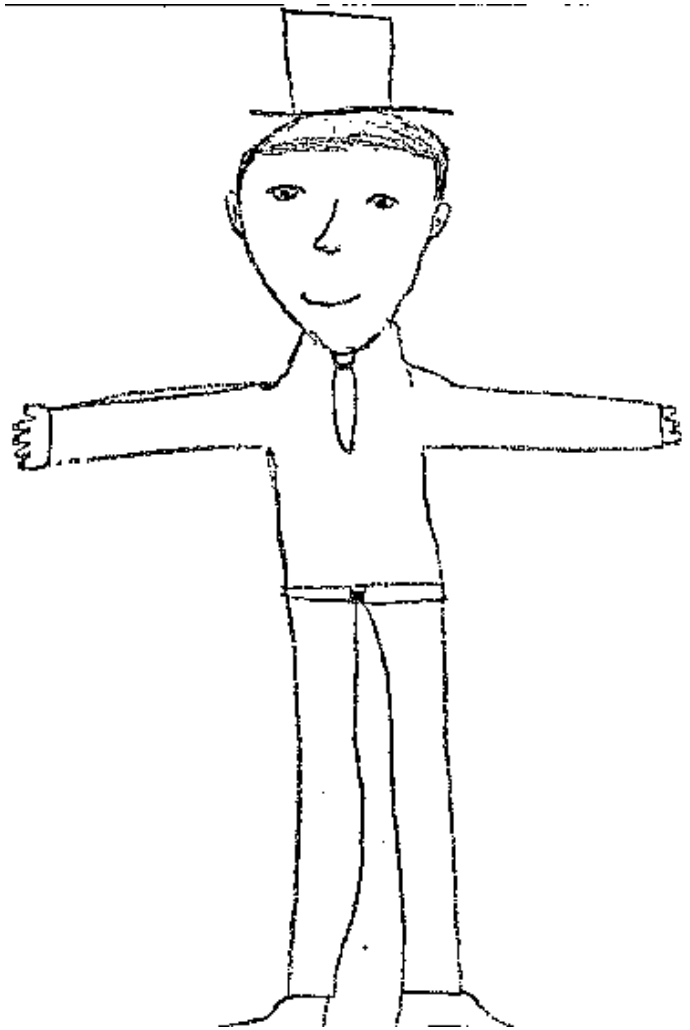
Age	Building with bricks	
15 months	Tower of 2 bricks	
18 months	Tower of 3 bricks	
2 years	Tower of 6 bricks	
2.5 years	Tower of 8 bricks or train with 4 bricks	
3 years	Bridge (from a model)	
4 years	Steps (after demonstration)	

Fine motor – Pencil skills

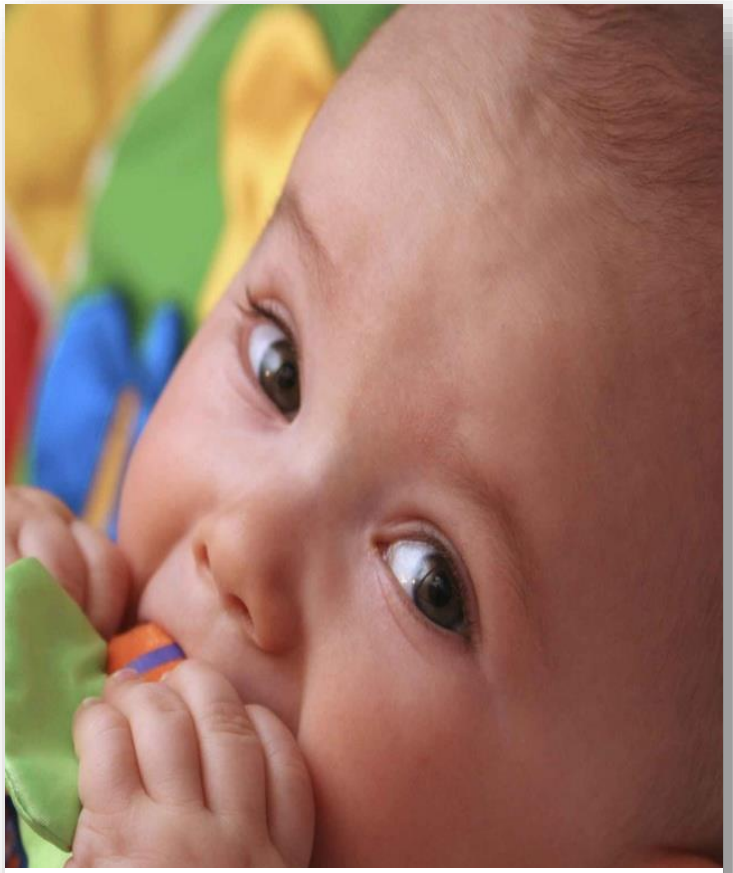
Age	Pencil skills
14 months	Scribbles with pencils
2 years	Copies a straight line
3 years	Copies a circle
4 years	Copies a cross
4.5 years	Copies a square
5 years	Copies a triangle Draws a person with 6 parts
6 years	Writes name

Draw man test

- **What is the developmental age ?**
  - 5-6 years or More than 5 years
- **Formula that you use**
  - 3+(n\4)
- **If child draw man with 20 parts what is expected age ?**
  - 8 years old



What is the developmental age for each picture ?



8 months

18 months

2 years

شرح

Hearing, Speech, & Language

Age	Hearing, Speech, & Language
Newborn	Startles to loud noise
3 months	Vocalizes, laughs and coo
6 months	Babbles ‘da’, ‘ma’
9 months	Dada, Mama
12 months	2-3 words other than dada/mama, Commands with gesture
15 months	4 to 6 words in addition to above, Uses jargon, Respond to one step verbal demand
18 months	10 single words (4) سنوات
21 months	use two or more words to make simple phrases
2 years	Vocabulary 50 words, 2-to-3-word phrases, Uses I and me (4) سنوات
3 years	Vocabulary 200 words, Uses 5-to-8-word sentences, Speech in sentences (4) سنوات
4 years	Count to 20 or more, Tells a story
5 years	Grammatical speech, Asks what words mean
6 years	Identifies written letters and numbers

Social, Emotional & Behavioral

Age	Playing	Age	Dressing & Potty
9 months	Plays gesture games Understands own name stranger anxiety	12 months	Cooperate with dressing
10 months	Waves bye-bye Plays “peek-a-boo”	2 years	Pulls off some clothing
18 months	Symbolic play, kisses parents, Plays near (but not with) other children	5 years	Dresses & undresses alone
3 years	Interactive play Takes turns	18 months	tells mother she/he wants potty
4 years	Imaginary play	2 years	Dry by the day
5 years	Plays cooperative games Understands rules and abides by them	4 years	Goes to the toilet alone Brush teeth Washes and dries hands

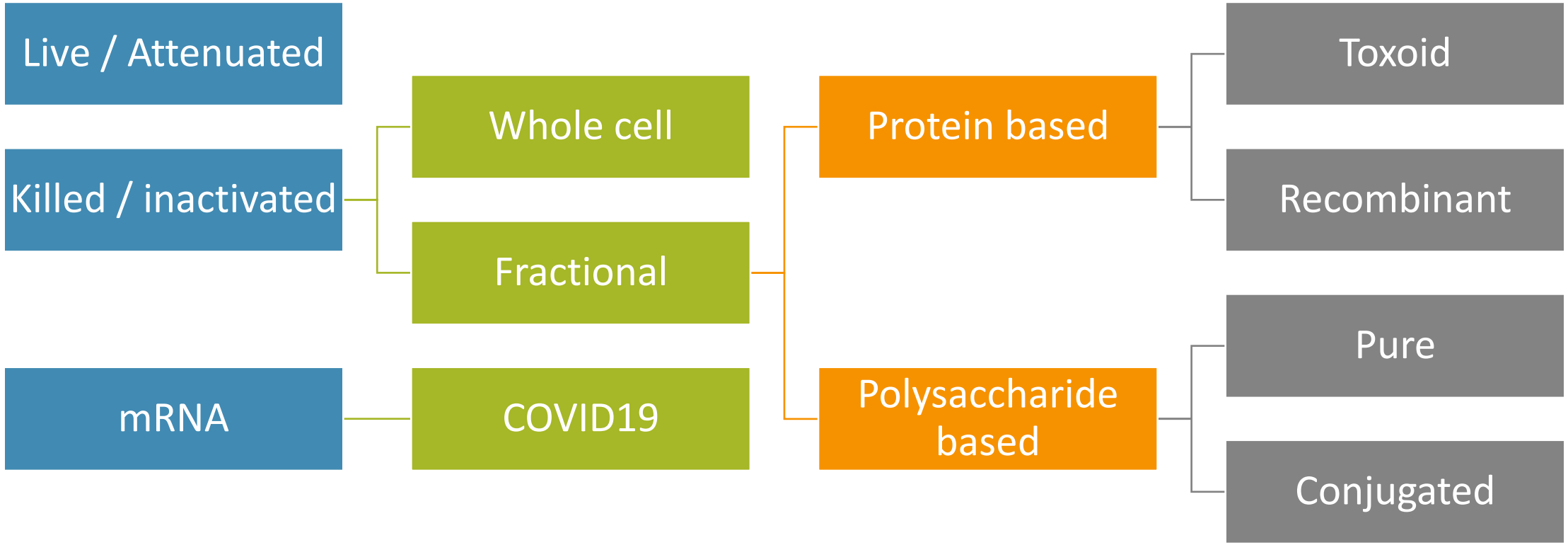
Immunization

Medium-Yield

Passive vs active immunity

	Passive	Active
MEANS OF ACQUISITION	Receiving preformed antibodies	Exposure to exogenous antigens
ONSET	Rapid	Slow
DURATION	Short span of antibodies (half-life = 3 weeks)	Long-lasting protection (memory)
EXAMPLES	IgA in breast milk, maternal IgG crossing placenta, antitoxin, humanized monoclonal antibody	Natural infection, vaccines, toxoid
NOTES	After exposure to tetanus toxin, HBV, varicella, rabies virus, botulinum toxin, or diphtheria toxin, unvaccinated patients are given preformed antibodies (passive)—"to Heal very rapidly before dying"	Combined passive and active immunizations can be given for hepatitis B or rabies exposure

Types of vaccines



Give example For Each Type Of Vaccines

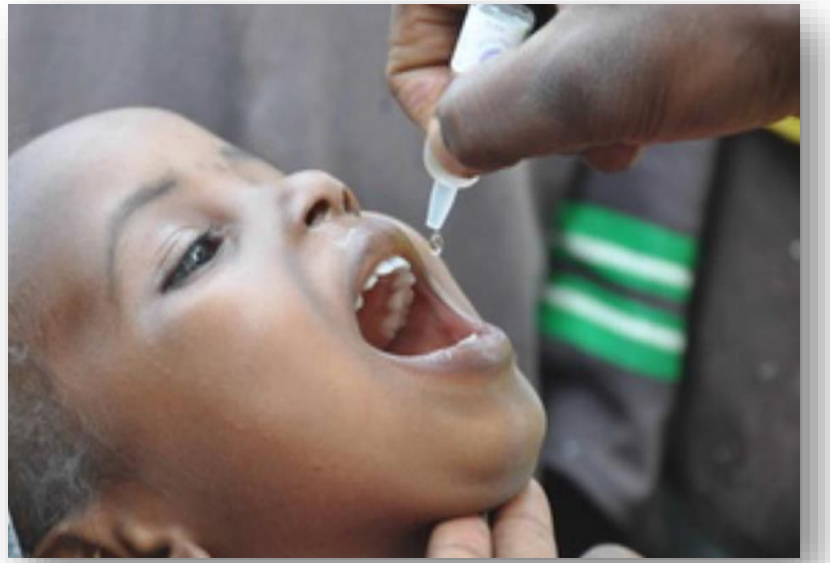
Live attenuated	Whole cell	Polysaccharide	Recombinant	Toxoids	mRNA
• BCG • MMR • OPV • Rota • Varicella • Yellow fever • Adenovirus • Smallpox	• Pertussis • IPV • Influenza (IM) • HAV • Rabies	• Hib • Neisseria meningitidis • Streptococcus pneumoniae	• HBV • HPV	• Tetanus • Diphtheria	• SARS-CoV-2

Route & Site of Administration for Vaccines

Intradermal	Subcutaneous	Intramuscular	Oral
BCG	Measles	HBV	Rota
The BCG vaccine is given intradermally only because if it's given subcutaneously, a local abscess may form that can sometimes ulcerate, and may require treatment with antibiotics immediately. Otherwise, without treatment, it could spread the infection causing severe damage to vital organs	Yellow fever	IPV	OPV
		DTwP, DTaP, DT, Td, TT	
		Hib	
		PCV-7	

Polio Vaccine

- **What do we call this vaccine ?**
  - OPV
- **What's the age of this child ?**
  - 91 days (& more)



Post-BCG vaccine abscess

- This baby took a vaccine. After 6 weeks he developed this lesion with axillary LNs enlargement

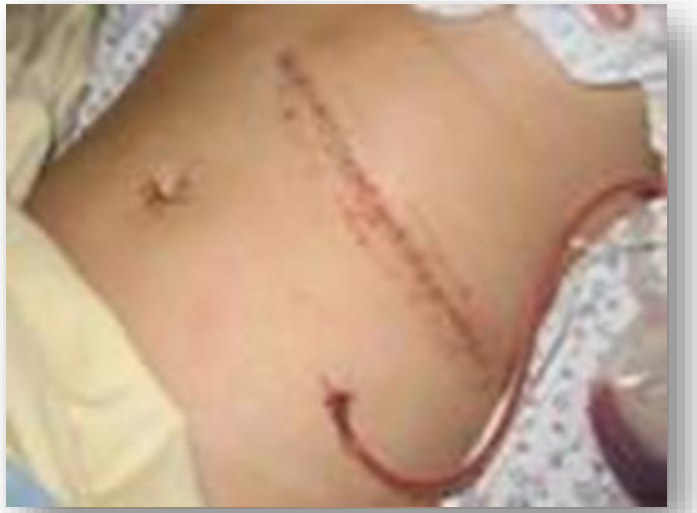
- **What is your spot diagnosis ?**
  - Post-BCG vaccine abscess formation with regional lymphadenitis.

- **What is the treatment ?**
  - Antibiotics

Side effects of the DTP vaccination

- **Mild Problems (Common):**
  - Fever, Redness, swelling, Soreness
  - Fussiness ,Tiredness or poor appetite and Vomiting
- **Moderate Problems (Uncommon):**
  - Seizure
  - Non-stop crying for 3 hours or more
  - High fever
- **Severe Problems (Very Rare):**
  - Serious allergic reaction
  - Long-term seizures, coma, or lowered consciousness
  - Permanent brain damage

6 YO child with this scar on his abdomen



- **What vaccines would you like to give him ?**
  - Give pneumococcal & meningococcal vaccines
  - Check Hib antibody titer to evaluate the need of a booster dose

- Post splenectomy patients should receive vaccines against encapsulated bacteria (SHiNS)
  - S.pneumonae
  - H.infleunzae
  - N.meningitidis
  - Salmonella; No vaccine is available right now

Vaccine adverse effects

	Local	Systemic	Allergic
Adverse effect	Pain, swelling, redness at site of injection	Fever & non-specific symptoms	Anaphylaxis Emergency treat with adrenaline
Rarity	Common	Common	Rare
Timing	Within 24-48h	DPT: 24-48h Measles 7-10 days	Within 5-30 min
Type of vaccine	Common with inactivated vaccines	Common with live attenuated vaccines	Any

إضافي

Contraindications

	General	Specific
Absolute	Anaphylaxis or a severe hypersensitivity reaction to a vaccine or a vaccine component should not receive subsequent doses of that vaccine	• DTP: History of Encephalopathy • Rotavirus: History of intussusception
Relative	Moderate or severe acute illness (can be vaccinated as soon as the recover)	**For DTP vaccine** • Progressive neurologic disorder • Temperature of 40.5° C or higher within 48h after previous DPT vaccine • Collapse or shock-like state within 48h after previous DPT vaccine • Seizure within 3 days after receiving a previous dose of DTP/DTaP • Persistent, inconsolable crying lasting 3 or more hours within 48 after previous DPT vaccine
Invalid	Recovery phase of acute illness, Recent exposure to infectious diseases, Current antibiotic therapy, Breastfeeding, Stable neurological condition, History of penicillin allergy or other non-vaccine allergy	

The Jordanian National Immunization Program

Age	Vaccine
Within 1st month	BCG Vaccine
2 months Day 61	Hexaxim vaccine (Hexa) - first dose, ROTA vaccine - first dose
3 months Day 91	Hexaxim vaccine (Hexa) - second dose, ROTA vaccine - second dose, OPV - first dose
4 months Day 121	Hexaxim vaccine (Hexa) - third dose, ROTA vaccine - third dose, OPV - second dose
9 months	Measles vaccine, OPV - third dose, Vitamin A (100 thousand international units)
12 months	MMR-1, HAV
18 months	MMR-2, DaPT, OPV, HAV, Vit A (200 thousand international units)
6 years	Td
15 years	Td

4 months

- **What vaccines you give for a 4 months old baby ?**
  - Hexaxim vaccine (DaPT-Hib-HBV-IPV) - third dose
  - ROTA vaccine - third dose
  - Oral Polio Vaccine (OPV) - second dose
- **What is the route of administration ?**
  - Hexaxim vaccine = IM
  - Rota, OPV = Oral

Source: https://www.unicef.org/jordan/stories/routine-childhood-immunizations

بس انتبهوا على شغلة نحن بنحكي هون على اليوم 121 هم باعتبروه 5 أشهر وهكذا فحتلاقوا المطاعيم نفسها لكن مكتوب على عمر أكبر بشهر من المحاضرة

A Nurse is giving injection to a 2-month-old baby

- **What is she giving him ?**
  - Hexaxim vaccine (DaPT-Hib-HBV-IPV) – first dose
  - At this age he is also given ORAL Rota vaccine

Source: https://www.unicef.org/jordan/stories/routine-childhood-immunizations

بس انتبهوا على شغلة نحن بنحكي هون على اليوم 61 هم باعتبروه 3 أشهر وهكذا فحتلاقوا المطاعيم نفسها لكن مكتوب على عمر أكبر بشهر من المحاضرة

If the baby take this vaccine, what is the estimated age ?

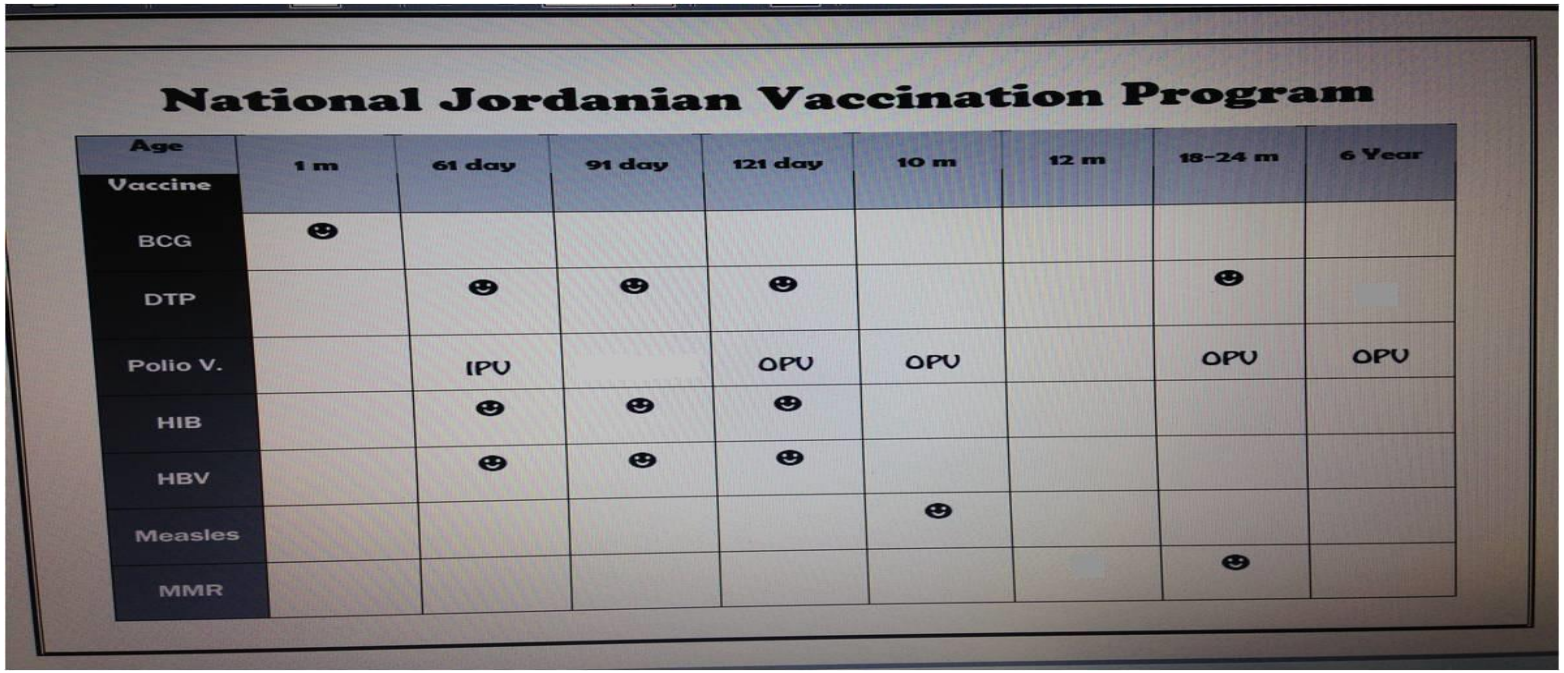
1. Measles vaccine
2. Oral Polio Vaccine (OPV) - third dose
3. Vitamin A (100 thousand international units)

❖9 months

What are the absent vaccine by time ?

السؤال قديم قبل إضافة مطعوم ال"Rota" ومطعوم ال"HAV"

- at 91 day … IPV and OPV
- at 12 month… MMR
- at 6 year… dT



National Jordanian Vaccination Program

Age / Vaccine	1 m	61 day	91 day	121 day	10 m	12 m	18-24 m	6 Year
BCG	☻							
DTP		☻	☻	☻			☻	
Polio V.		IPV		OPV	OPV		OPV	OPV
HIB		☻	☻	☻				
HBV		☻	☻	☻				
Measles					☻			
MMR							☻	

Growth chart

High-Yield

Stature

Short stature

- Below 3rd percentile for age and sex
- **Causes**:
  1. Genetic familial
  2. Down syndrome
  3. Malnutrition
  4. Hypothyroidism
  5. Constitutional growth delay
  6. Growth hormone deficiency
  7. Any chronic disease as CF

Tall stature

- Above 97th percentile for age and sex
- **Causes**:
  1. Genetic familial
  2. Acromegaly
  3. Klinefelter syndrome
  4. Collagen disorder as Marfan

إضافي

Head circumference

Microcephaly

- Below 3rd percentile for age and sex
- Seen with
  1. Genetic familial
  2. Down syndrome
  3. Phenyl ketonuria
  4. Congenital infections
  5. Craniosynostosis
  6. Osteogenesis imperfecta

Macrocephaly

- Above 97th centile for age and sex
- Seen with
  1. Genetic familial
  2. Congenital hydrocephalus
  3. Congenital diseases

Failure To Thrive

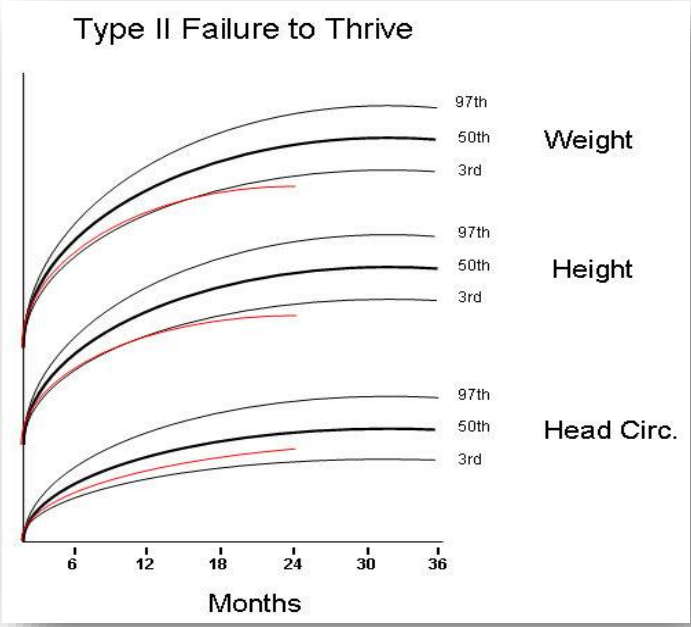
- **Defined as 1 of the following**
  - Fall over 2 or more percentile
  - Are persistently below the third or fifth percentiles
  - Are less than the 80th percentile of median weight for height measurement

- **Stunting**: Low height for age

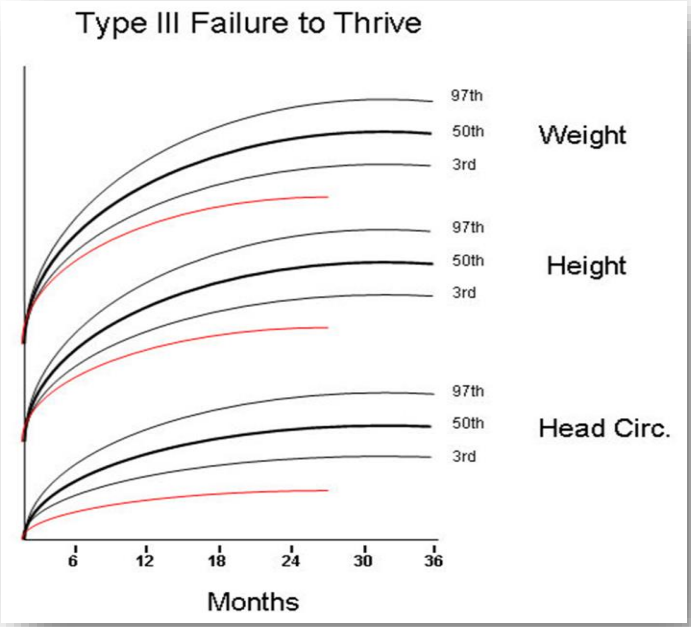
- **Wasting**: Low weight for height

Types of Failure To Thrive

- Inadequate intake of calories
- Inadequate caloric absorption
- Excessive caloric requirements



- Familial short stature
- Constitutional growth delay
- Hypothyroidism, growth hormone deficiency, hypopituitarism
- And chronic malnutrition

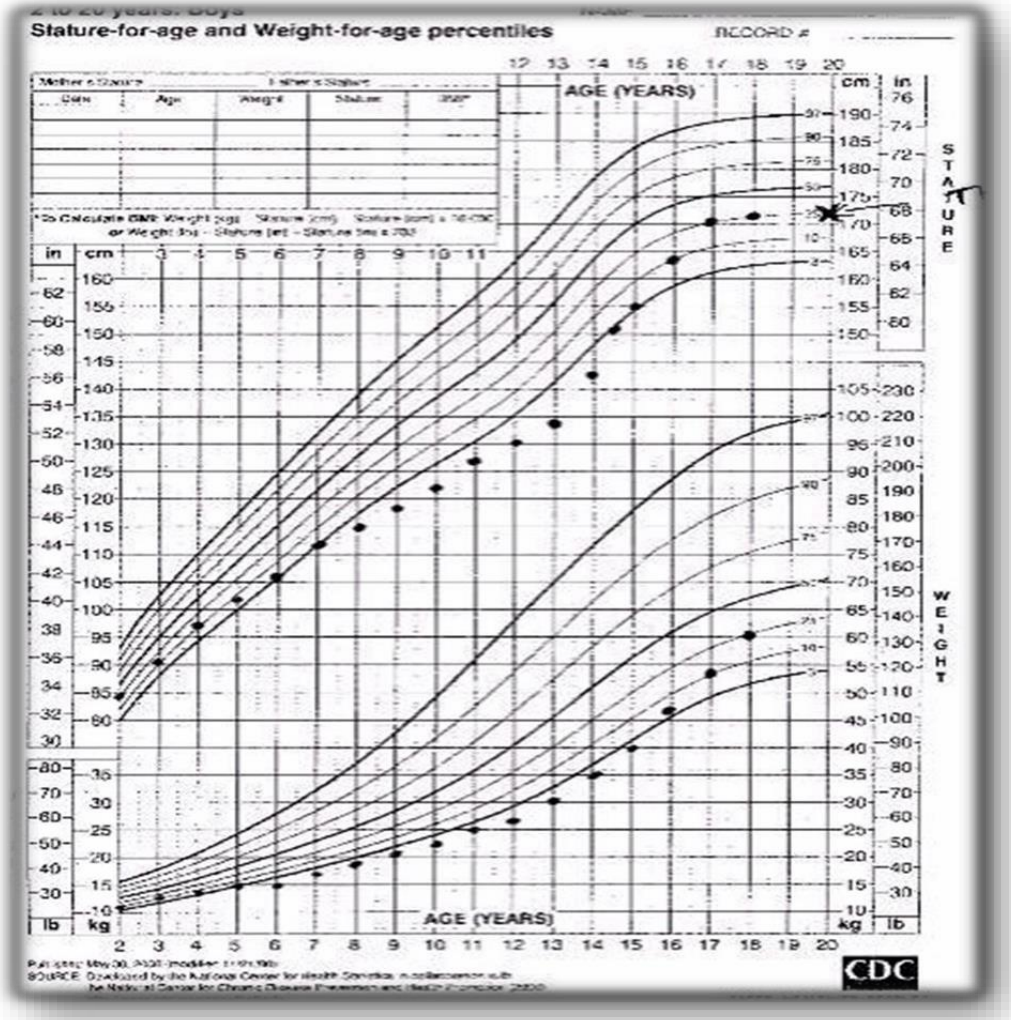


This growth pattern often begins at and usually results from:

- Intrauterine infections
- Chromosomal abnormalities
- Prenatal exposure to toxins

Growth chart

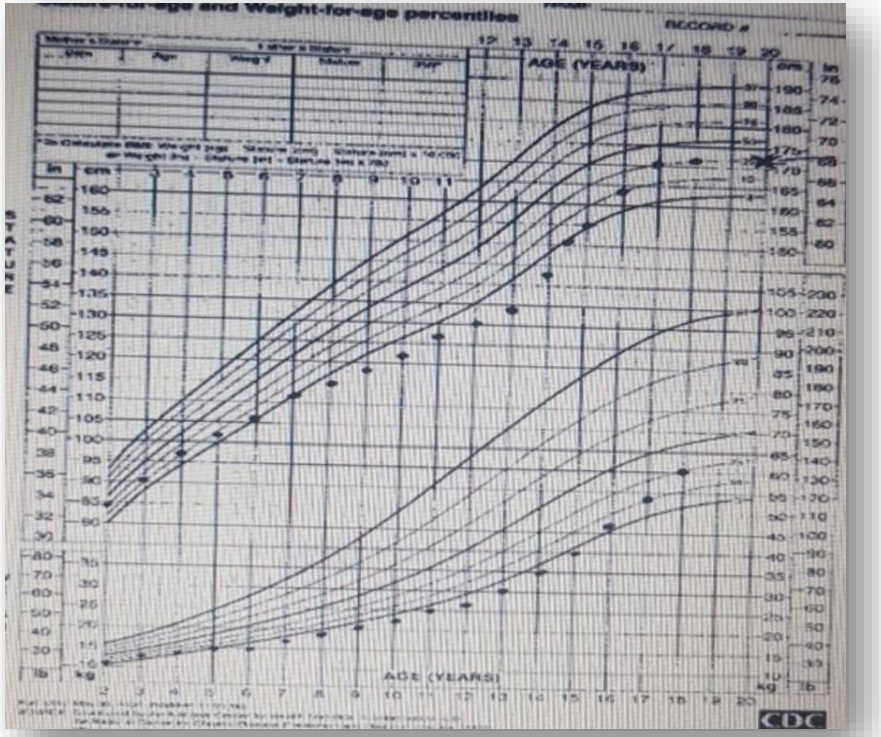
- This chart for patient not receiving any treatment
- **What is the cause behind his growth delay?**
  - Constitutional delay growth



- This chart for a healthy child
- **What is the cause behind his growth delay?**
  - Constitutional delay growth

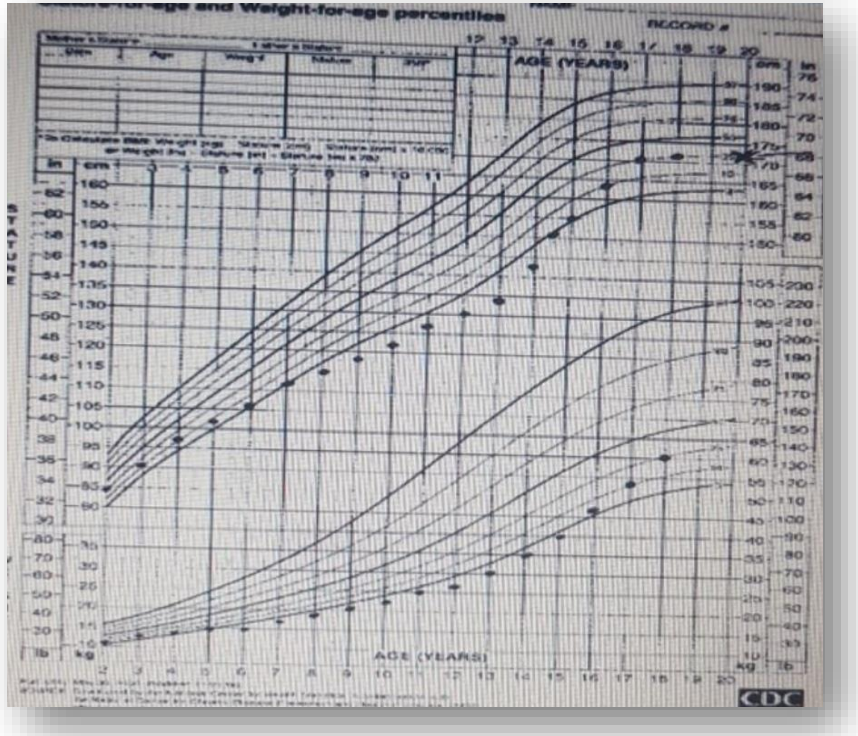
Growth chart

- **What is the abnormality in this chart ?**
  - Short stature
- **If this girl's mother had her first period at 14 years, what is the cause of her problem ?**
  - Constitutional delay
- **How to confirm diagnosis ?**
  - By wrist x-ray
- **What is the treatment ?**
  - No treatment needed, just wait



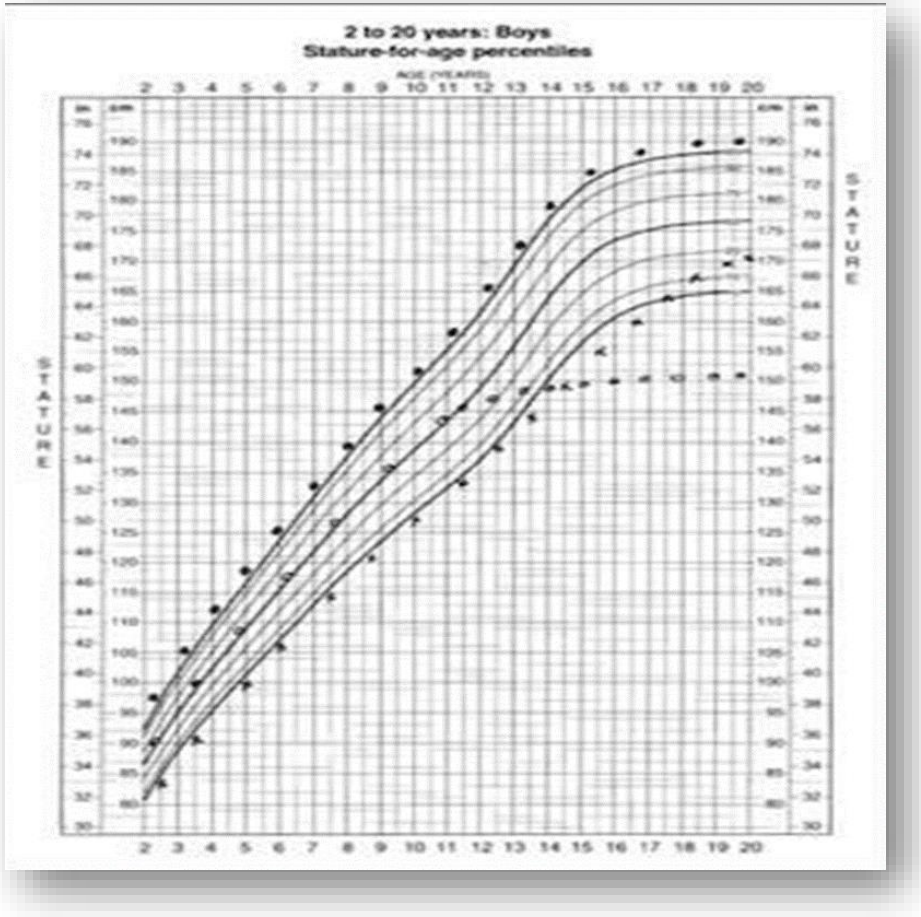
Based on this growth chart, the most likely relevant diagnosis is

a. Genetic short stature
b. Chromosomal abnormality
c. Constitutional growth delay
d. Growth hormone deficiency
e. Congenital hypothyroidism



Growth chart of 3-year-old boy

- **What's the abnormality in this Growth chart?**
  - Short stature
- **What's the most appropriate Dx.?**
  - Celiac disease.



Growth chart

- 12-year-old male complained of chronic diarrhea, with positive family history of DM type 1. the growth chart of patient presented

❖ **What is the abnormality at growth chart?**

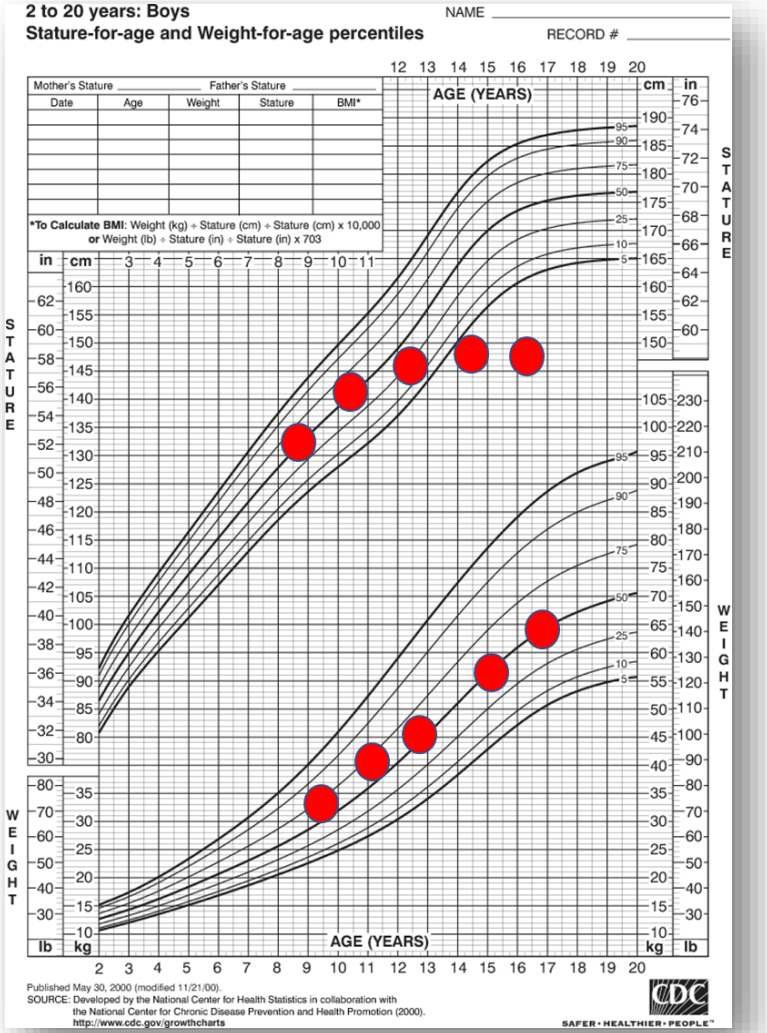
- Short stature

❖ **What is the most sensitive test for diagnosis of that patient?**

- Anti-tissue glutaminase and endomysial IgA abs

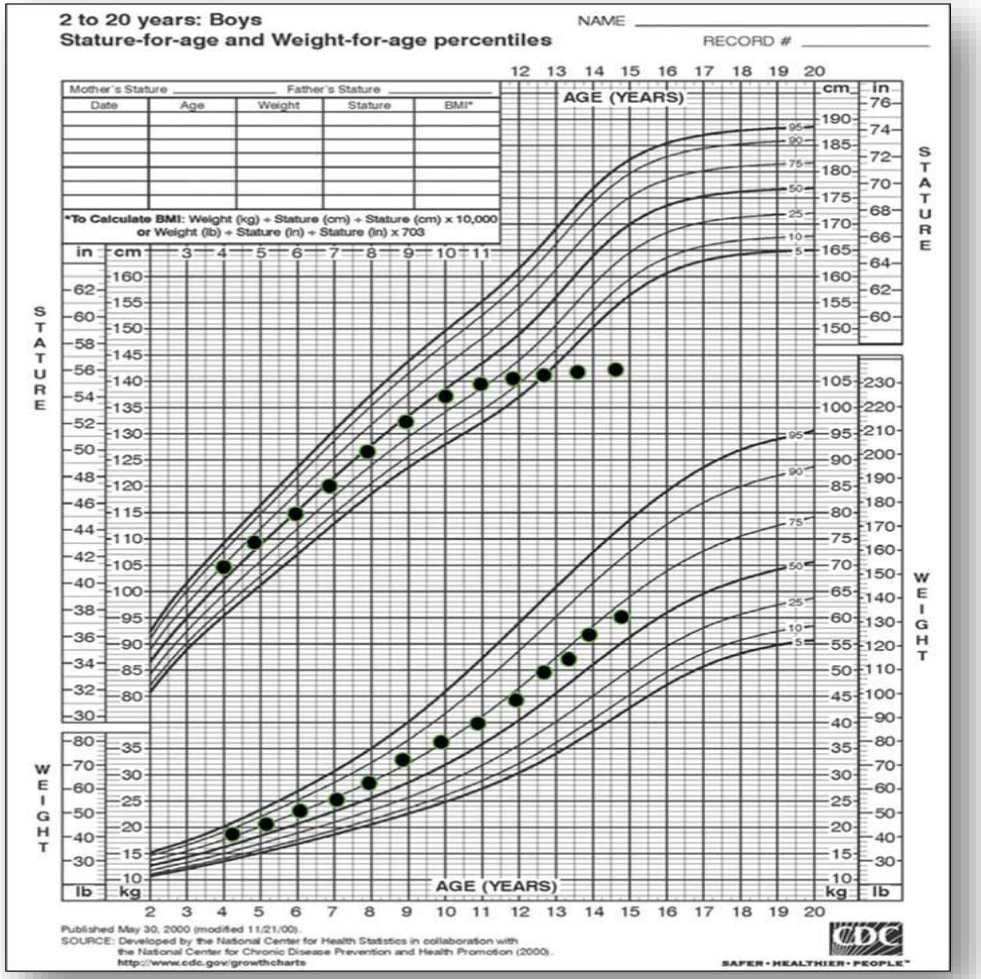
❖ **What is the treatment?**

- Gluten free diet



Growth chart

- 15-year-old female presented with short stature, she also has constipation, skin changes and neck swelling
- **What is the most likely diagnosis ?**
  - Hypothyroidism
- **What tests you'd like to order ?**
  - TSH, T3, T4 levels
- **What is the treatment?**
  - Levothyroxine



Growth chart

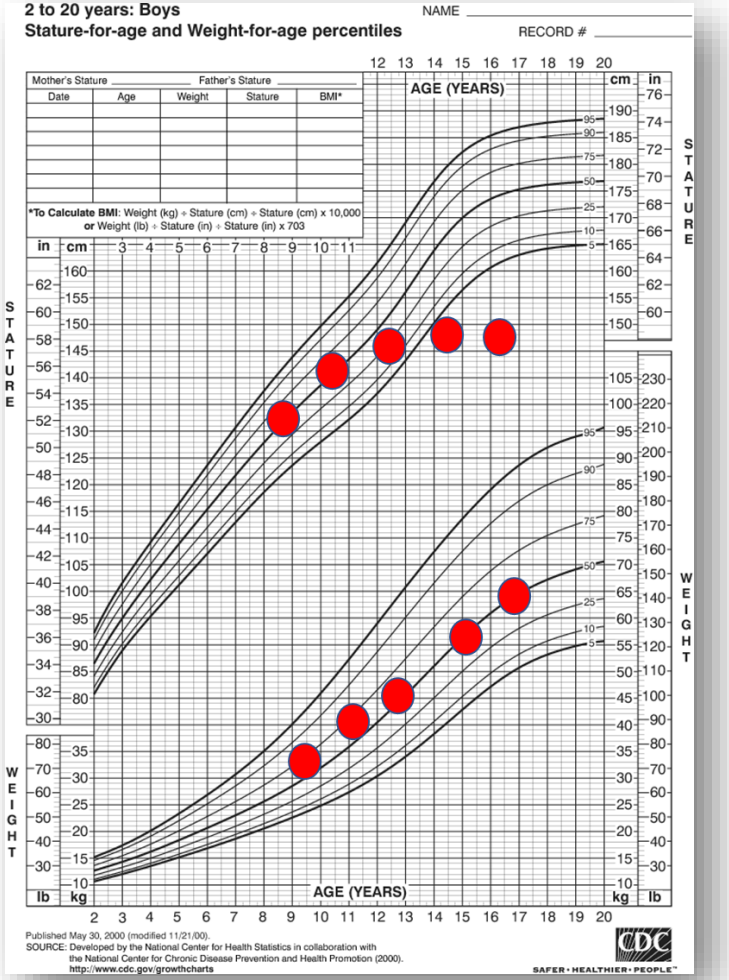
- 12-year-old with positive family history of DM type 1. the growth chart of patient presented

❖ **If the patient complained of chronic diarrhea, what is the most likely diagnosis ?**

- Celiac disease

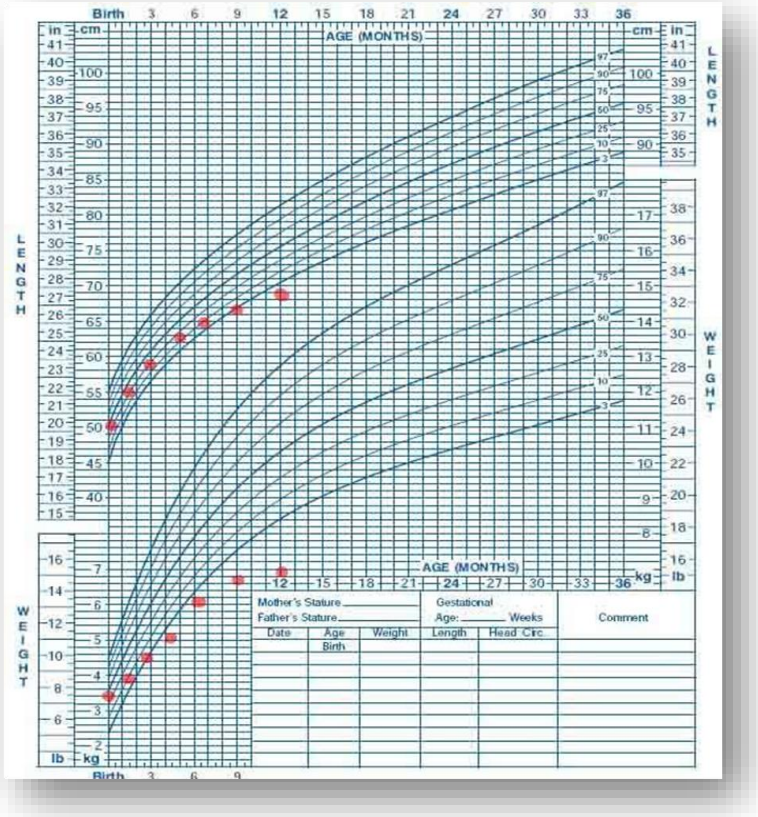
❖ **If the patient complained of chronic constipation and neck swelling, what is the most likely diagnosis ?**

- Hypothyroidism



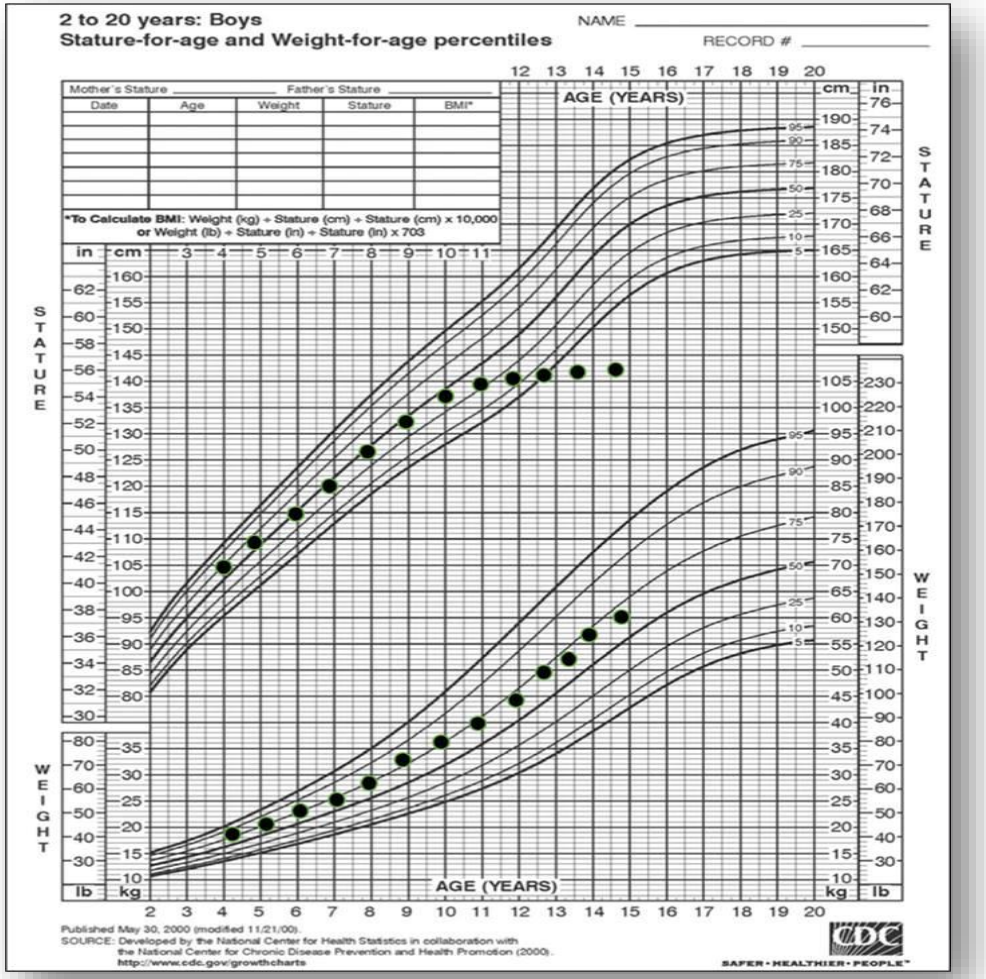
Growth chart

- 7-year-old male complained of chronic diarrhea, his sister was diagnosed with Hashimoto disease. the growth chart of patient presented.
- **What is the abnormality at growth chart?**
  - Short stature , weight for age that falls below the 5th) FTT
- **What is the diagnosis ?**
  - Celiac disease
- **What is the treatment?**
  - Gluten free diet



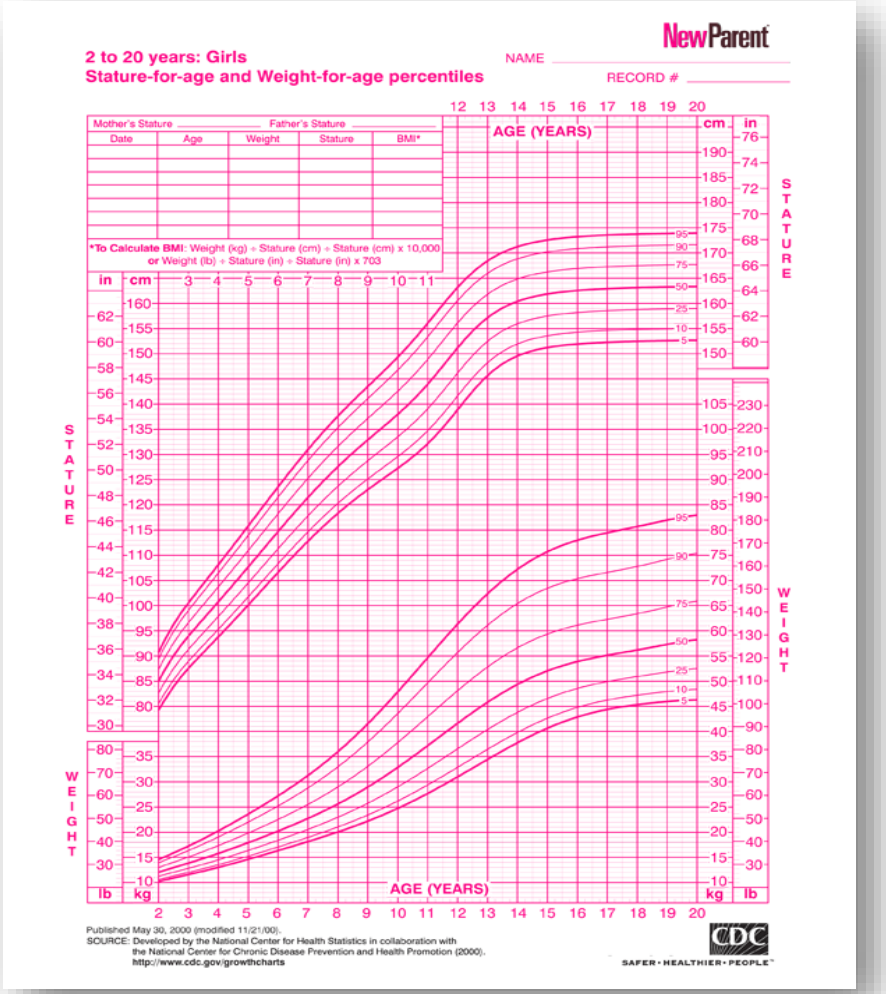
Growth chart

- **What is the abnormality ?**
  - Short stature
- **Mention 2 DDx**
  - Celiac disease
  - Hypothyroidism
  - Familial (genetic)



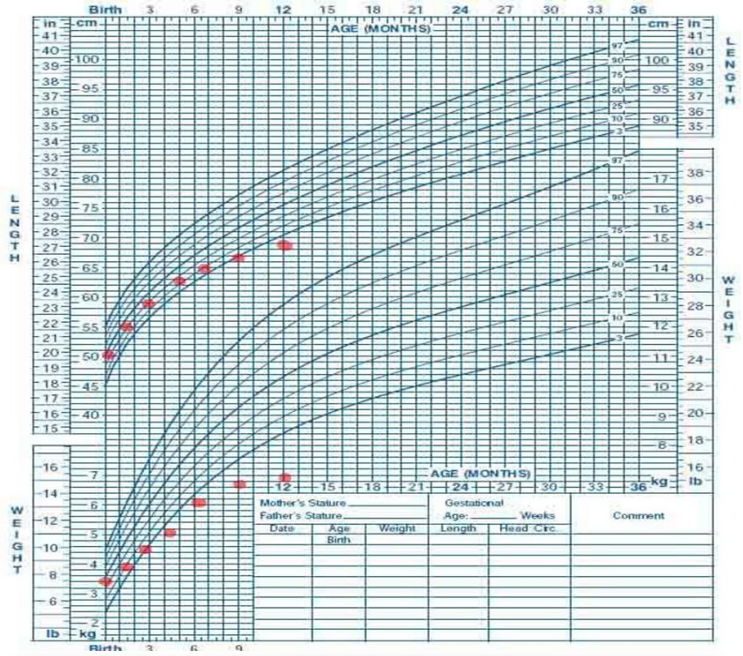
Growth chart

- According to the growth chart: 12-year-old female, weight =35kg, height=135cm
- **What are the findings?**
  - Weight = between the 10th and the 25th centile
  - Height = below the 5th centile/short stature
- **Give 2 possible causes**
  - Hypothyroidism
  - Celiac disease
  - Familial (genetic)



Growth chart

- **What is your diagnosis ?**
  - Cystic fibrosis
- **What tests you'd like to order ?**
  - Sweat chloride
  - Genetic study



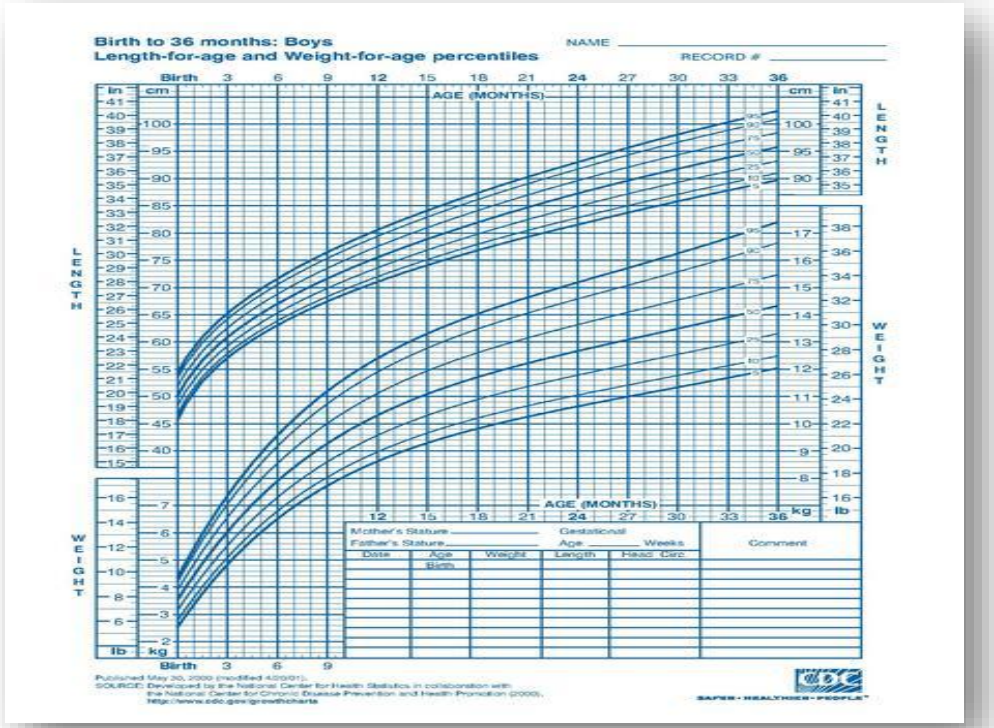
Growth chart

- 12 M/O male child his wight 6 kg and length 60 cm

- **What's the centile for his wight and length ?**
  - Both are under th 5th centile

- **What's the name of this condition ?**
  - FTT type 2

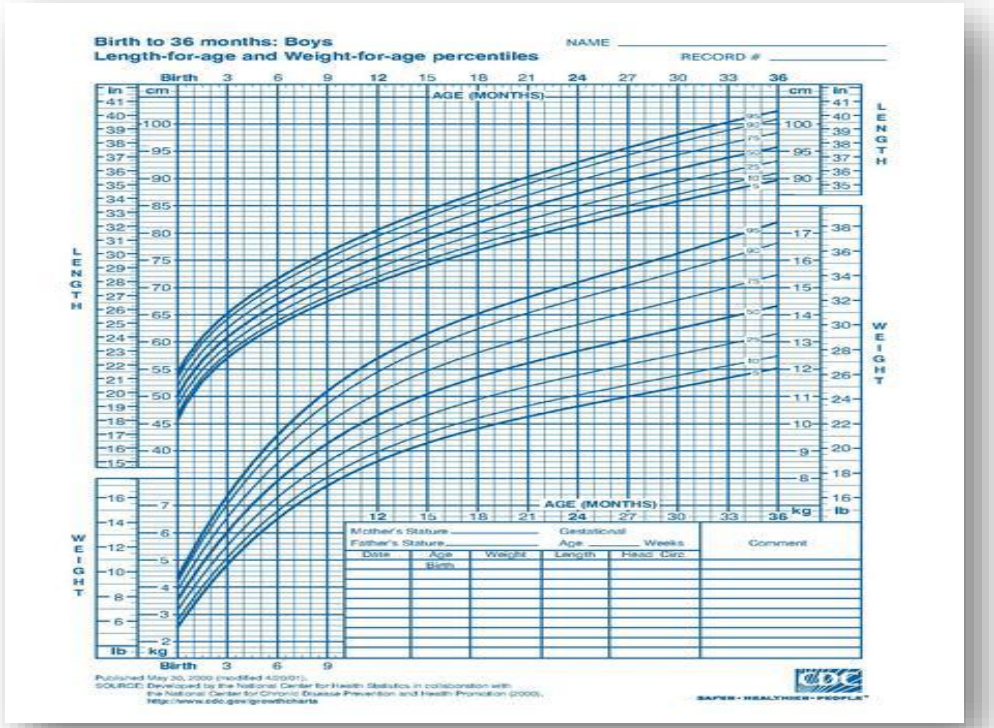
- **If his condition ass with recurrent chest infection what's the dx?**
  - Cystic fibrosis



Growth chart cont.

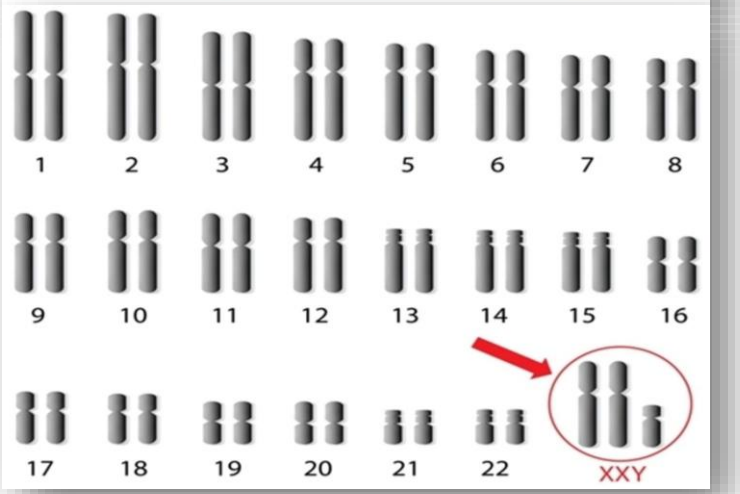
- 12 M/O male child his wight 6 kg and length 60 cm

- **How can you confirm your dx ?**
  - Sweat chloride more than 60 twice or once with positive genetic testing
- **According to national vaccine program. What's the vaccines should be given to this age ?**
  - MMR HAV
- **What extra vaccines are needed ?**
  - Pneumococcus and meningococcus



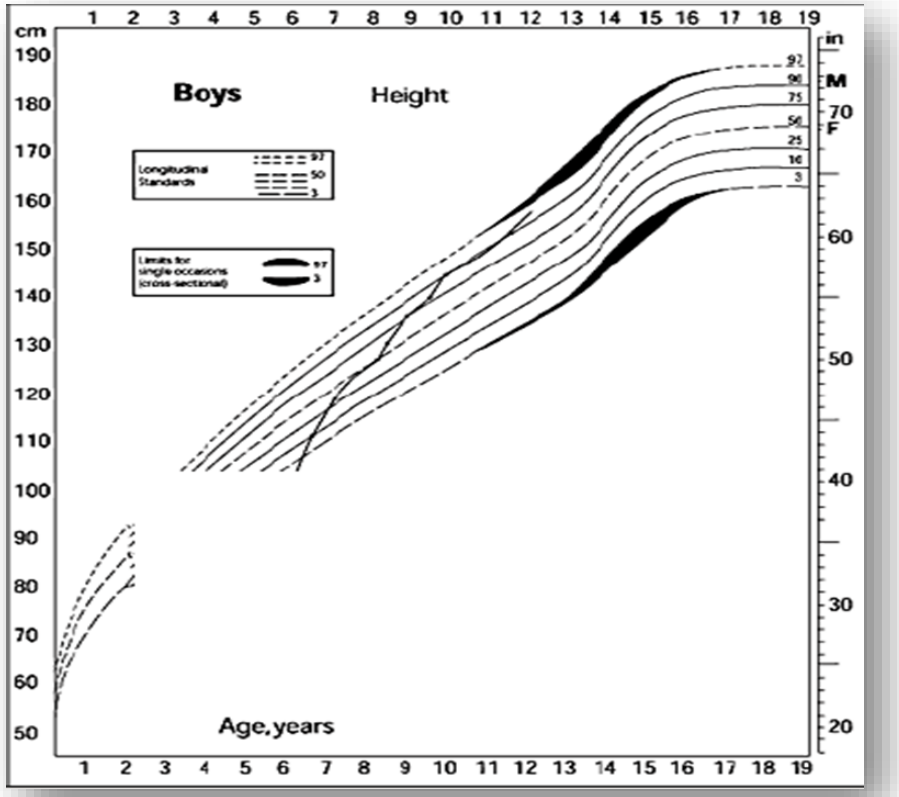
Growth chart

- According to the growth chart: 12-month-old male, weight = 12.5 kg, height= 83 cm
- **At which percentile is the height & weight ?**
  - Weight = between the 90th and the 95th centile
  - Height = above the 95th centile/tall stature
- **According to the growth chart and the karyotype what is your diagnosis ?**
  - Klinefelter Syndrome



Growth chart

- **What is the cause of the change in the growth of this child at the age of 8 (catch up) ?**
  - Receiving Growth hormone



Growth charts – BMI

- 4 years old girl weight 18 kg and her height 100 cm

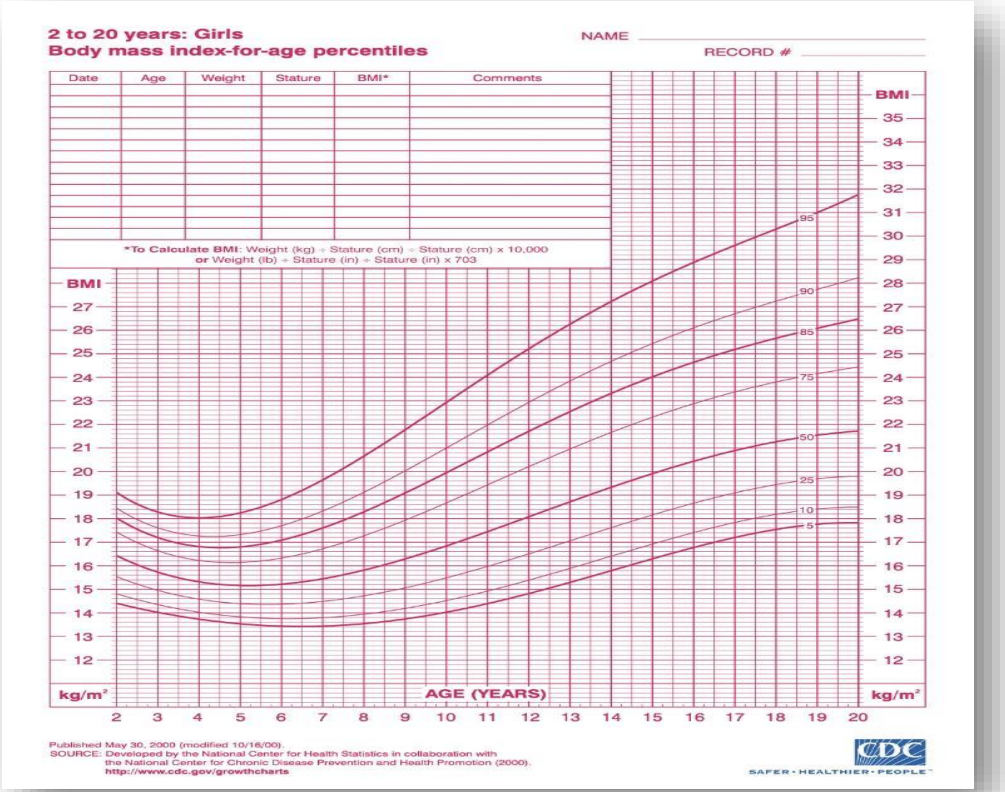
❖What is her BMI ?

- 18 kg/m^2

❖Interpretation on BMI chart ?

- Between 90 and 95 percentile
- She is overweight

Weight Category	Body Mass Index	
	Children	Adults
Underweight	Below 5th percentile*	Below 18.5
Healthy weight	5th percentile to less than 85th percentile	18.5 to 24.9
Overweight	85th percentile to less than 95th percentile	25 to 29.9
Obese	95th percentile or above	30 or above



Growth charts – BMI

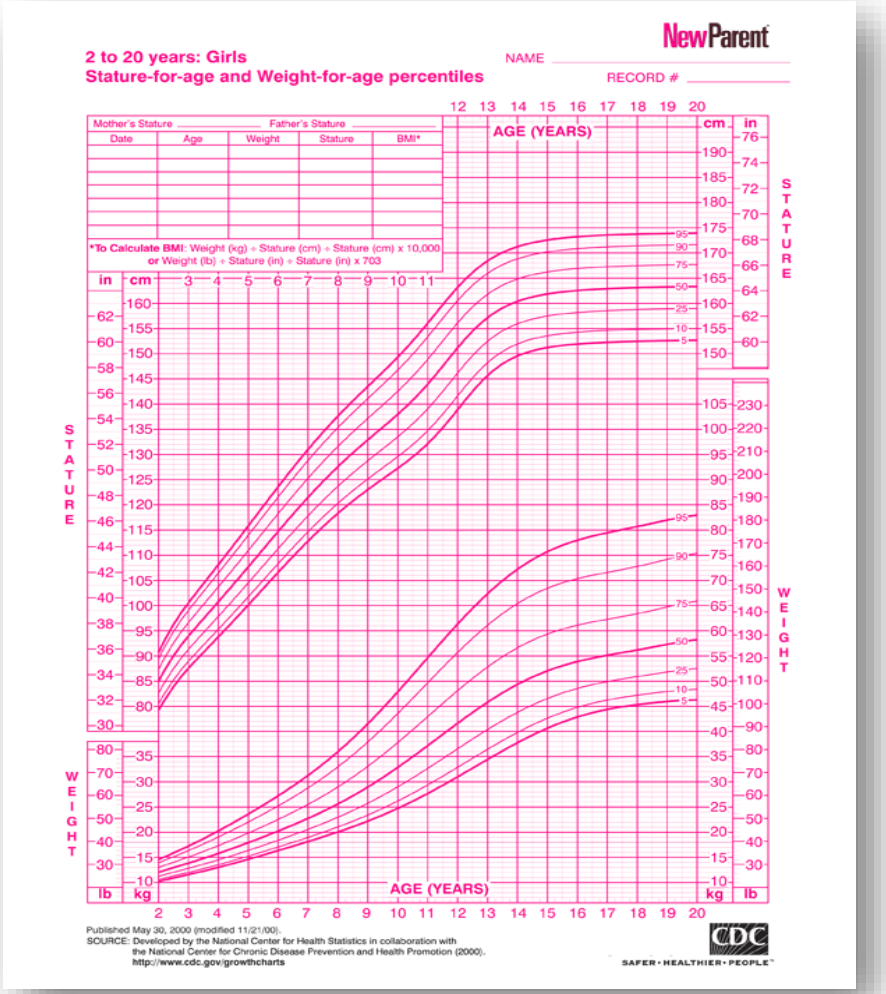
- 4 years old girl weight 20 kg and her height 105 cm

- **Interpretation on growth chart ?**
  - Her height is between 75th & 90th percentile
  - Her weight is on 95th percentile

- **What is her BMI ?**
  - 18.1

(use the chart on previous slide)

- **Interpretation on BMI chart ?**
  - Between 90th and 95th percentile
  - She is overweight

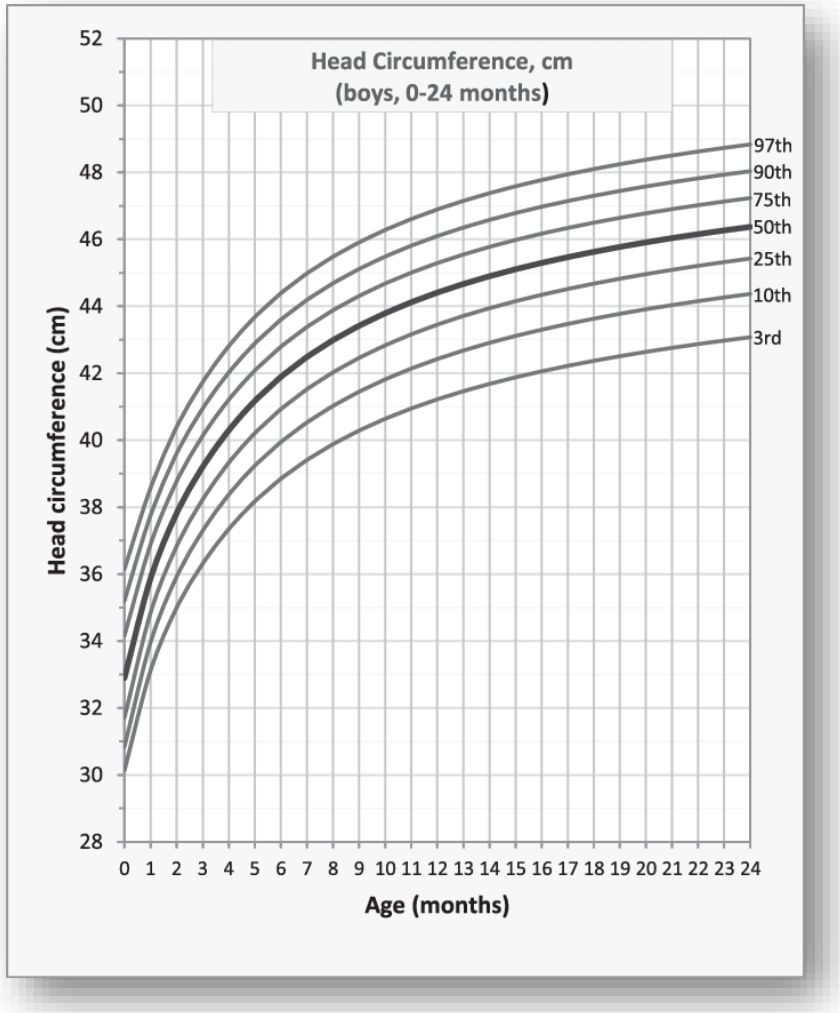


Growth charts – Head circumference

- The growth of 3-month-old boy, full-term, vaginal delivery, his head circumference is 36 cm now

- **What is the clinical diagnosis**
  - Microcephaly

- **Give 2 causes for your diagnosis**
  1. Genetic familial
  2. Down syndrome
  3. Phenyl ketonuria
  4. Congenital infections
  5. Craniosynostosis
  6. Osteogenesis imperfecta



Growth charts – Head circumference

- 10-month-old boy with head circumference 48

❖ **What's the centile ?**

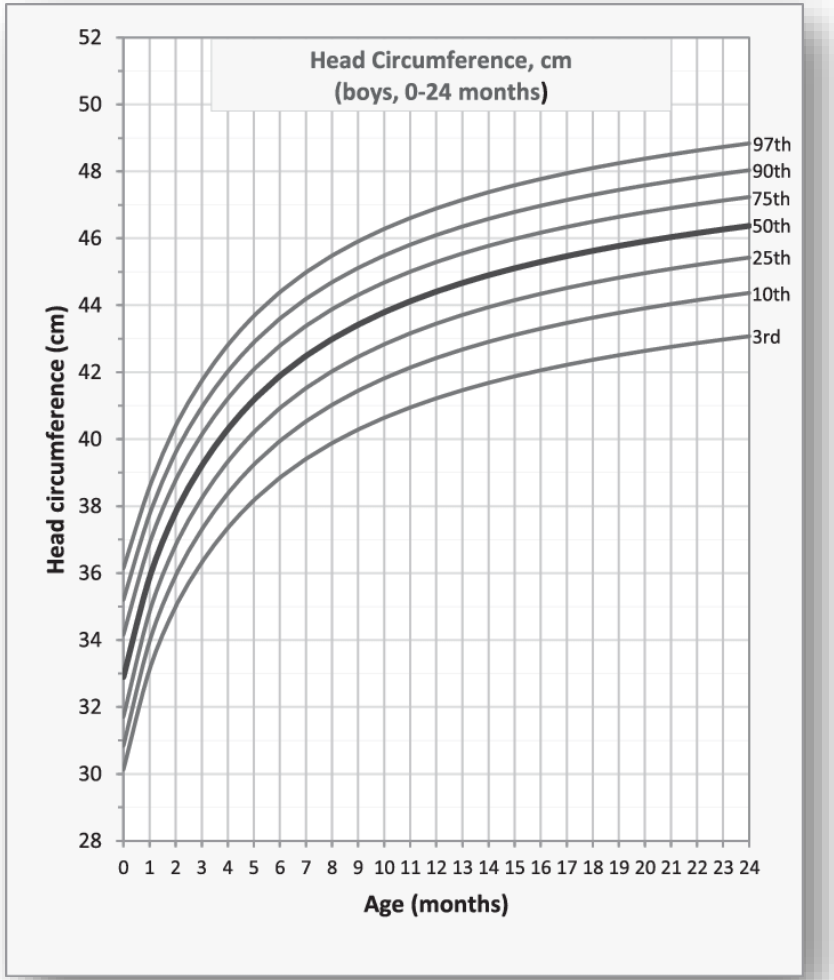
- Above 97th centile

❖ **What's the diagnosis ?**

- Macrocephaly

❖ **Give 2 causes for your diagnosis**

- Genetic familial
- Congenital hydrocephalus
- Intracranial tumor
- Hurlar syndrome
- Congenital hypothyroidism



Growth charts – Head circumference

- 2-months-baby with head circumference 42

❖What's the centile ?

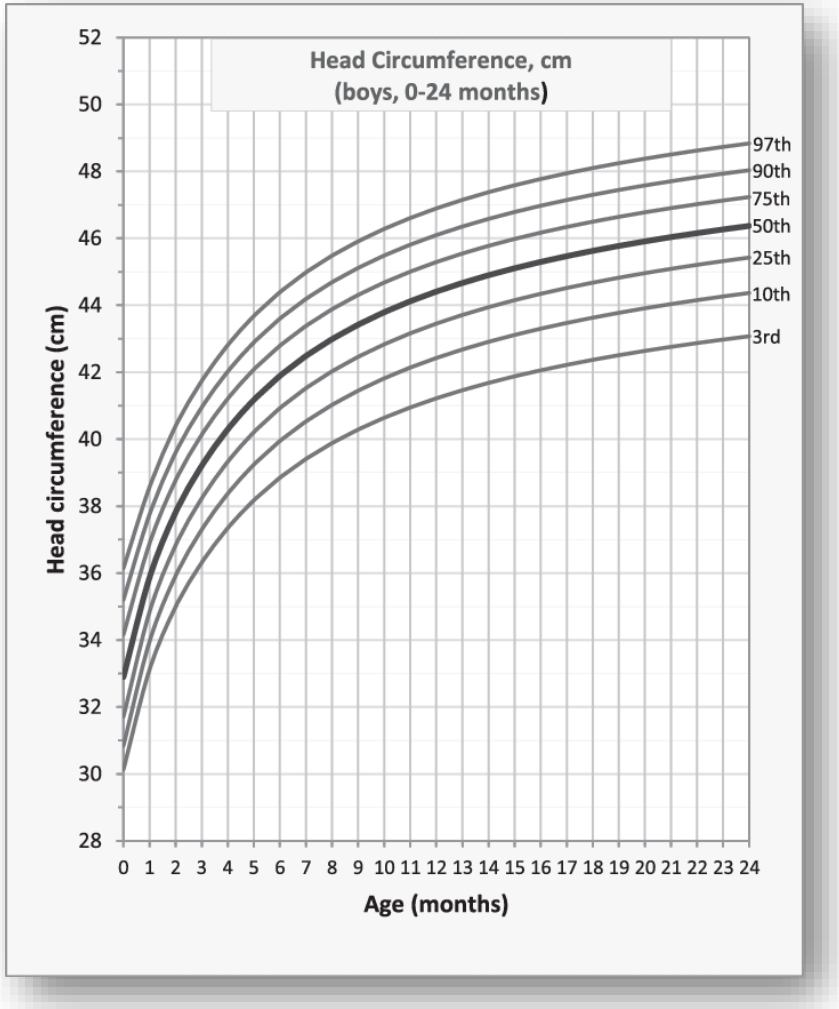
- Above 97th centile

❖What's the name of the sign ?

- Macrocephaly

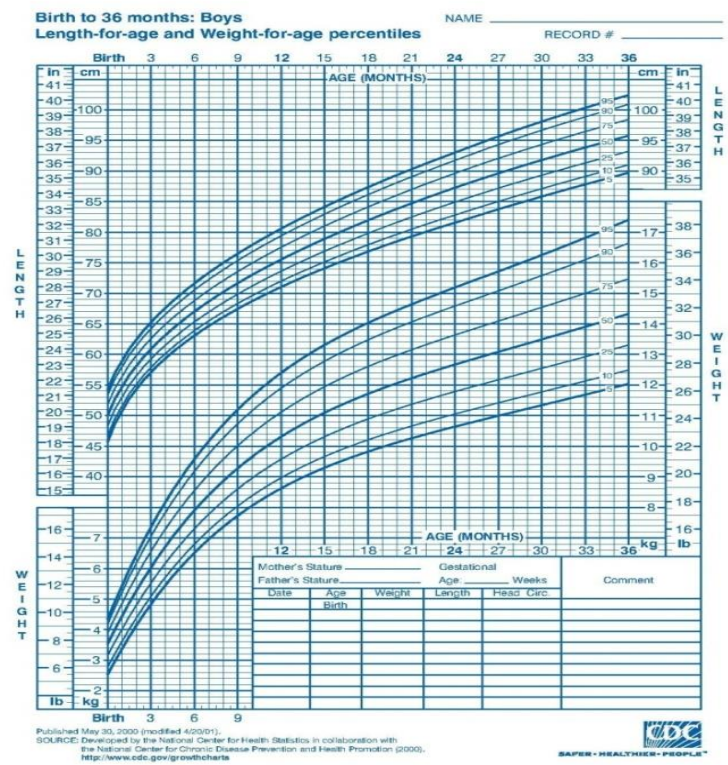
❖Give 2 causes for your diagnosis

- Genetic familial
- Congenital hydrocephalus
- Intracranial tumor
- Hurlar syndrome
- Congenital hypothyroidism



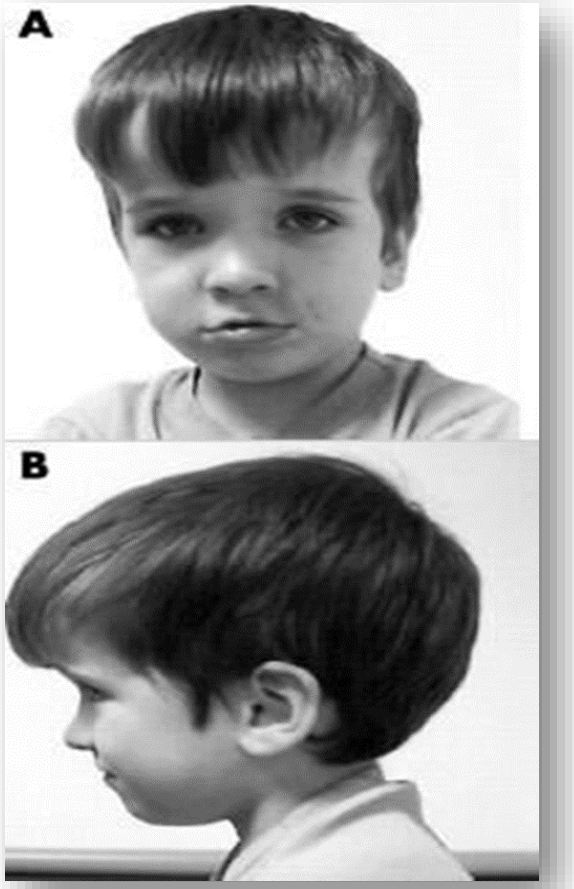
Growth charts – Head circumference

- 9-month-old patient, HC > 97th percentile
- **Give 2 DDx**
  - Genetic familial
  - Congenital hydrocephalus
  - Intracranial tumor
  - Hurlar syndrome
  - Congenital hypothyroidism
- **Mention one therapeutic measure**
  - VP shunt



Growth charts – Head circumference

- Growth chart of head circumference to age, the HC at 11th month became above 97th centile
- **What's your comment (what's that called)?**
  - Macrocephaly
- **What's the most likely cause of this presentation?**
  - Hydrocephalus
- **Mention 2 signs**
  - Sun set eyes, Papilledema
- **Mention 2 symptoms**
  - Vomiting, Headache



Infant formula

Medium-Yield

Breastfeeding

❖ **Mention WHO recommendation for breastfeeding Initiation within the 1st hour of life**

1. Exclusive breastfeeding (that is the infant only receives breast milk without any additional food or drink, not even water).
2. Breastfeeding on demand (that is as often as the child wants, day & night).
3. No use of bottles, teats or pacifiers.

Infant formula

Category	Example	Special indication
Cow's milk-based formulas	S26, Nan, Saha, Similac, bebelc, AR formulas, "Sensitive" / LF	–
Soy formulas	Isomil, ProSobee, Saha -LF	Galactosemia Lactase deficiency
Casein hydrolysate formulas	Babylac HA, Nan HA, Alfare (LF), Alimentum, Prigistamil	Cow's milk protein allergy
Amin acids-based formula (elemental)	Neocate, Elcare	Cow's milk protein allergy not responding to Casein hydrolysate formulas

*Purple: Not available in Jordan and is unlikely to come in exams

Infant formula

❖**What is the type and indication for each formula ?**

A. Anti-regurgitation (starch based), GERD

B. Amino acid based, cows milk protein allergy not responding to Casein hydrolysate formulas

C. Soy formulas, Galactosemia & Lactase deficiency

A

B

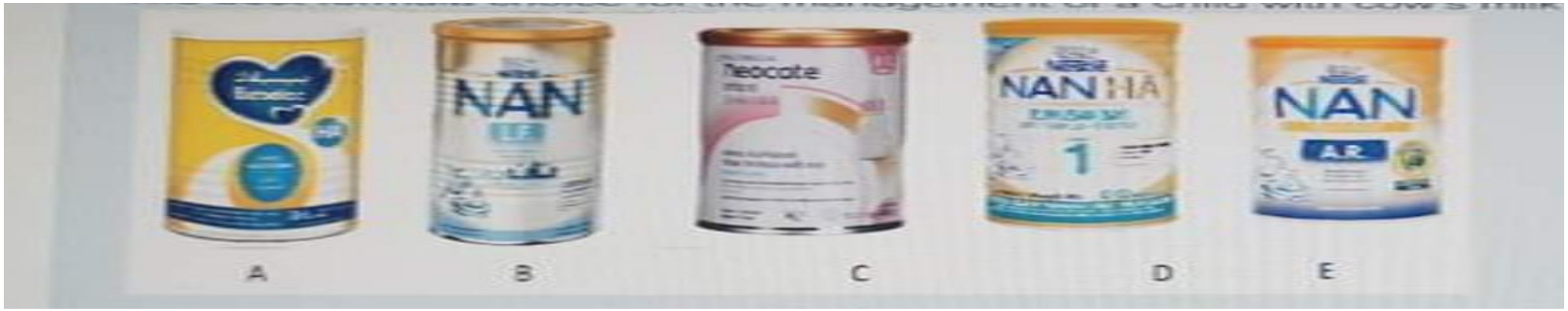
C

Infant formula

- **Mention the indication for each**
  - **L.F.**: Lactase deficiency
  - **A.R.**: GERD

Infant formula

- **The best formula used for patient with cow milk protein allergy ?**
  - Amino acid based; formula C
- **The best formula to use for the patient with GERD ?**
  - Anti-regurgitation (starch based) ; formula E

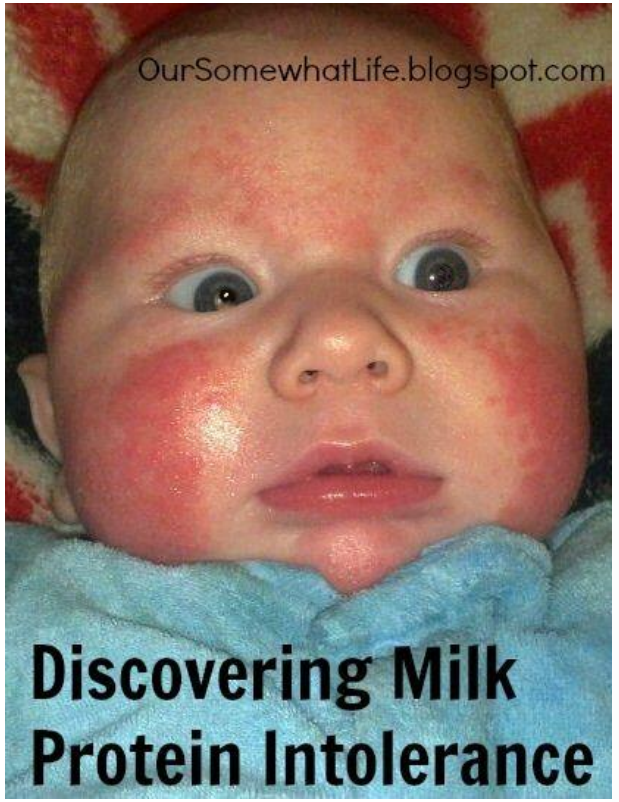


Cows milk allergy

- Type 1 hypersensitivity reaction or type 4 hypersensitivity reaction
- **Symptoms**:
  - Chronic diarrhea, vomiting, abdominal pain (after feeding), crying and irritability, blood in the stool (intestinal)
  - Extra intestinal: wheezy chest due to laryngospasm (reactive airway disease), skin rash, eczema, iron deficiency anemia, may lead to anaphylactic shock
- **Diagnosis**: High eosinophilia, Challenge elimination test
- **Treatment**: Casein Hydrolysate formula, Amino acid-based formula

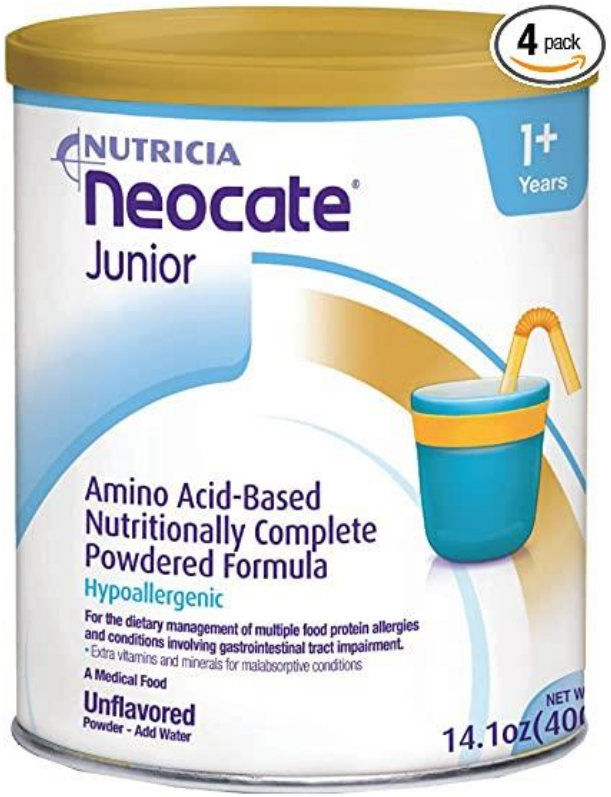
Infant on babylac formula and has diarrhea

- **What is the diagnosis ?**
  - Cows milk allergy
- **Finding in the stool?**
  - Eosinophils
- **What is the treatment ?**
  - Casein Hydrolysate formula
  - Amino acid-based formula



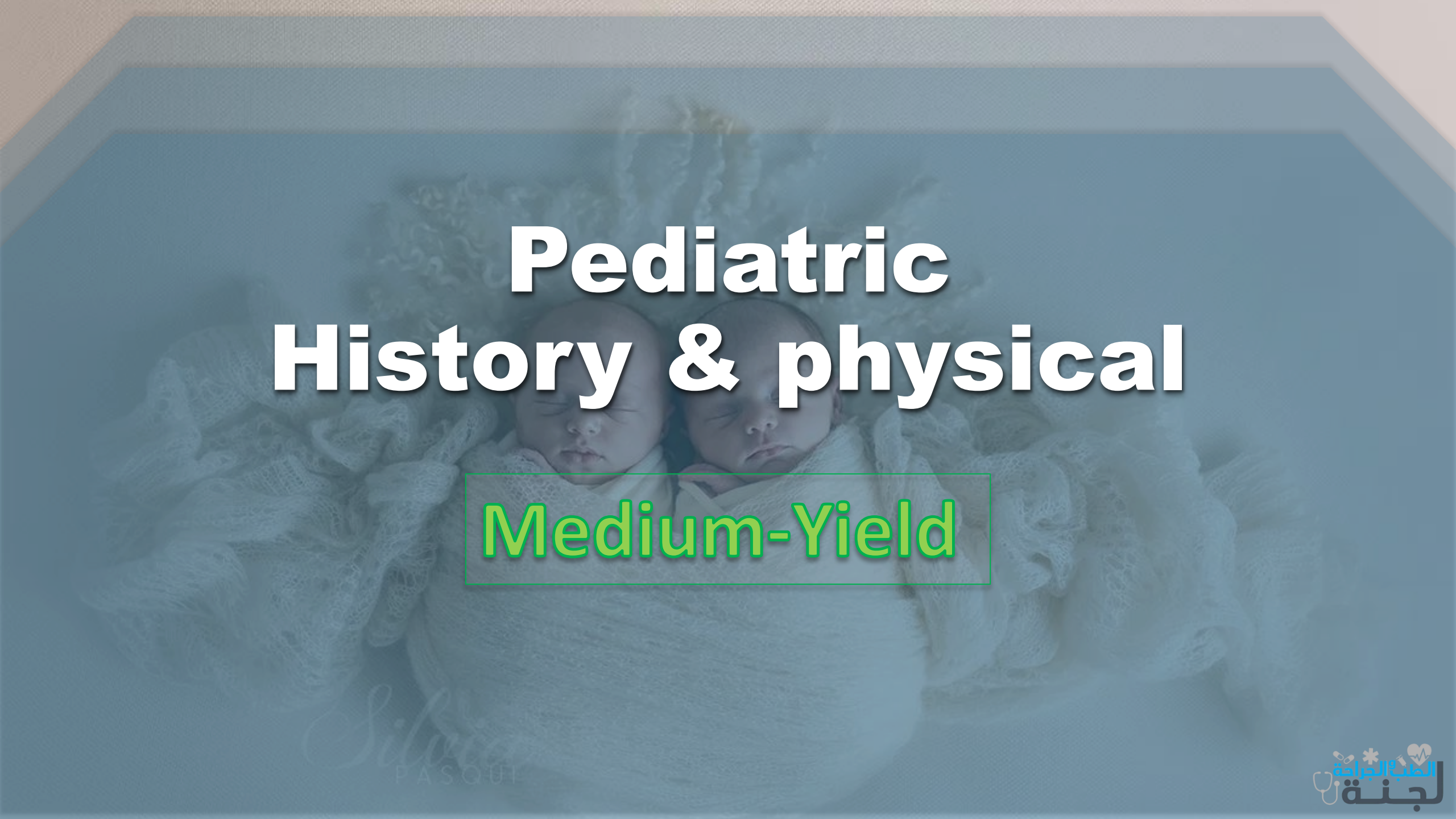
Cows milk allergy

- **What stool finding in a child who uses this formula?**
  - Eosinophils and RBC
- **Alternative formula?**
  - Hydrolyzed formula
- **Complication of this condition?**
  - Failure to thrive



Pediatric History & physical

Medium-Yield



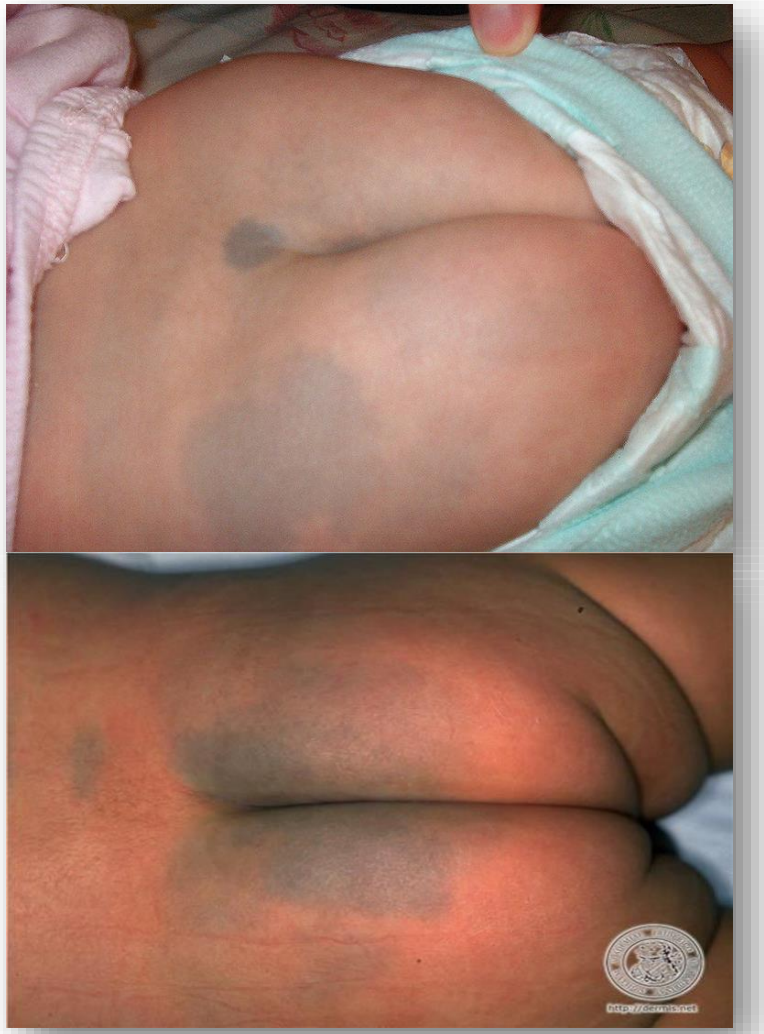
Mongolian spots

❖ **What's the Dx.?**

- Mongolian spots.

❖ **What you want to do for this patient, why?**

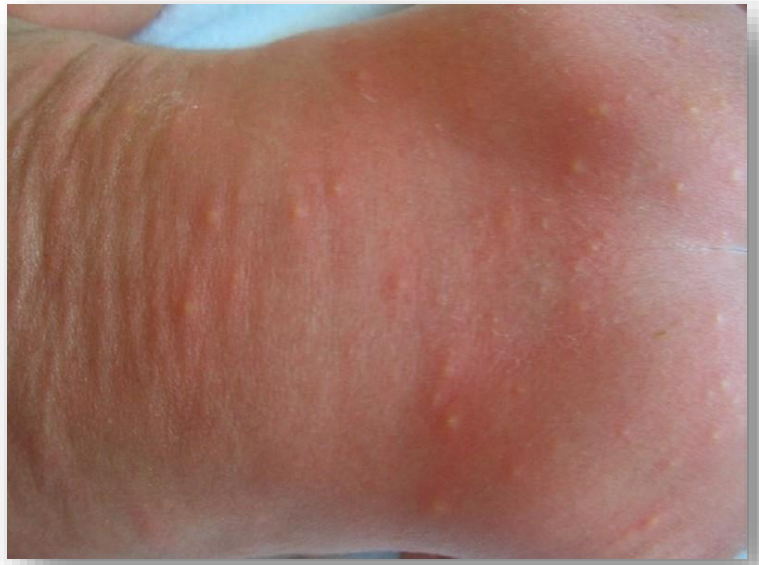
- Nothing, because it normally disappears 3–5 years after birth.



➢ **Notes**:

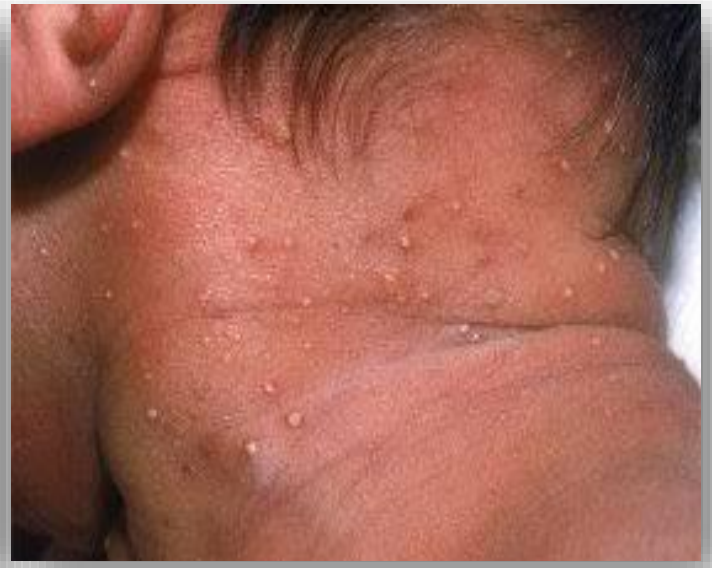
- Common in dark skin races
- Can occur anywhere, but mostly in the lumbosacral area
- Hypersensitive melanocytes to MSH
- For few months, then spontaneously resolve

Skin findings

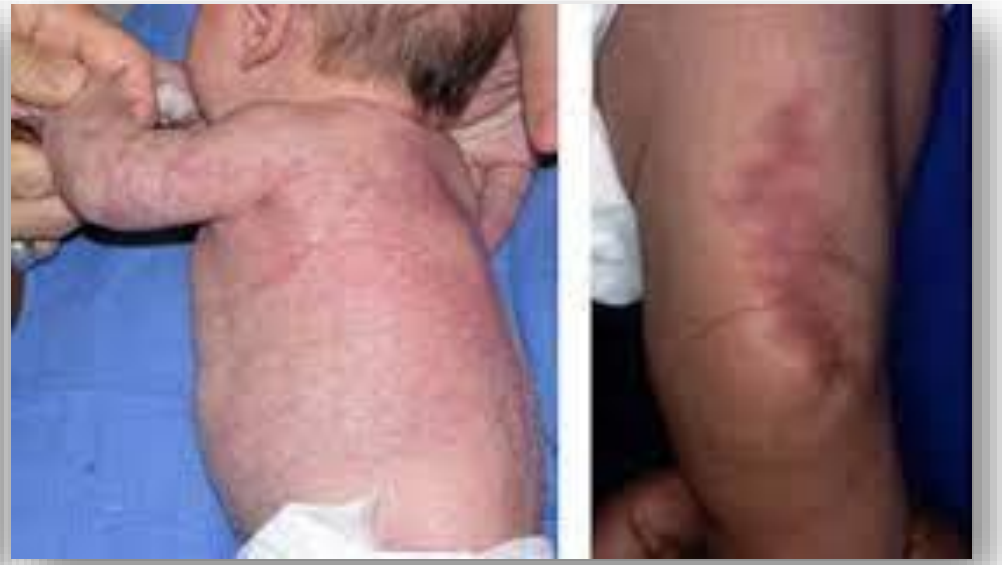


Erythema toxicum

- Not present in the first 24 hrs of life
- Appear as erythematic rash which will develop pustules
- Resolve within weeks



Pustular melanosis

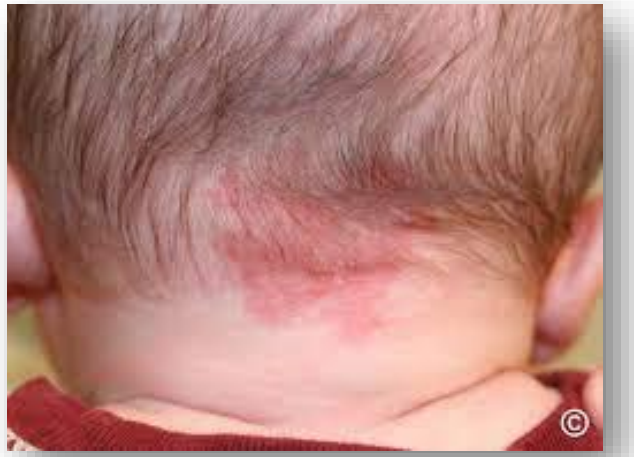


Cutis marmorata

- Called modeling of the skin
- Normal variant in newborn
- Seen with temperature problems
- Common with down syndrome

Skin findings

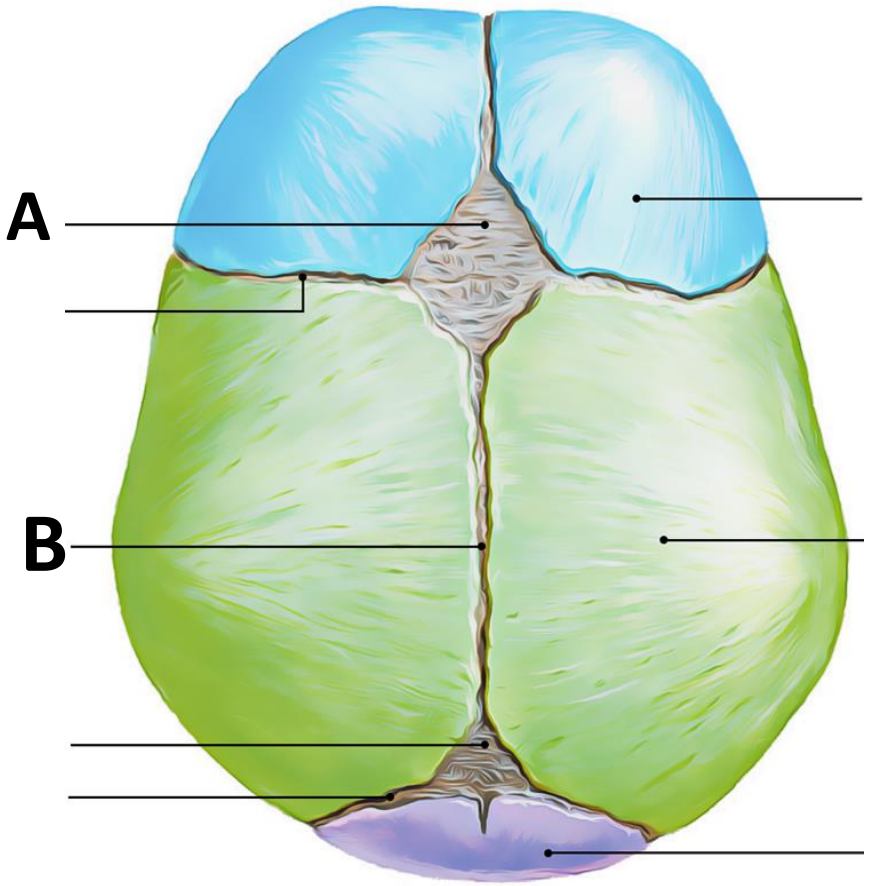
- **Salmon patch**
  - Very common in the back of the neck
  - Can also seen in face , eyelid
  - When the baby cry, it gets more red
  - Usually goes within weeks



- **Port wine stain**
  - Do not go away
  - Associated with Sturge weber syndrome (discussed earlier)
  - Its unilateral in the distribution of the trigeminal nerve

Fontanels

- **Mention 2 causes of wide structure A**
  - Rickets, congenital hypothyroidism, ..
- **Mention 2 signs seen with of depressed ant fontanel**
  - Sunken eye, tachycardia, skin turgor, …
- **What is the result of early closure of structure B**
  - Craniosynostosis >> Dolicho/Scapho-cephaly



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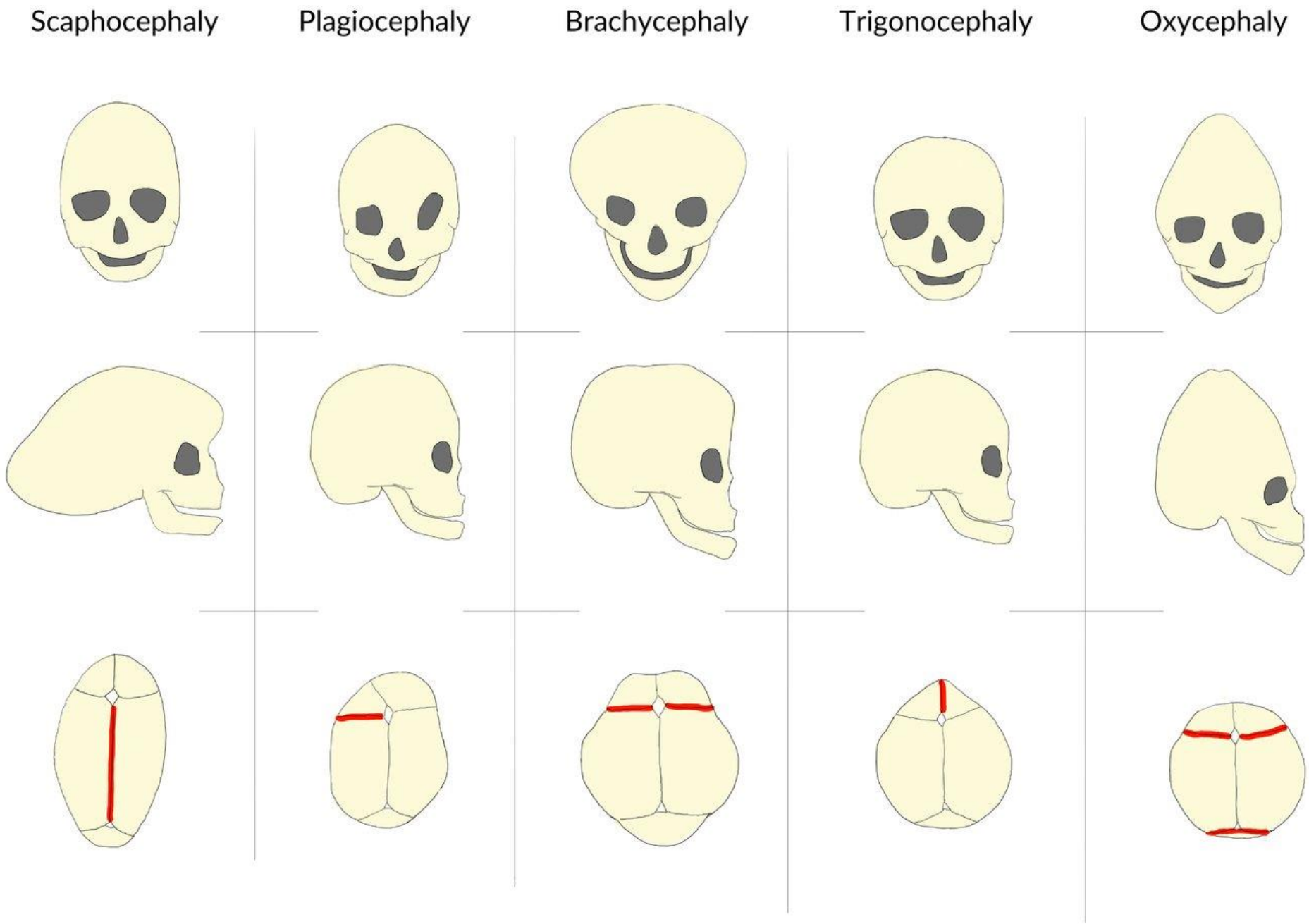
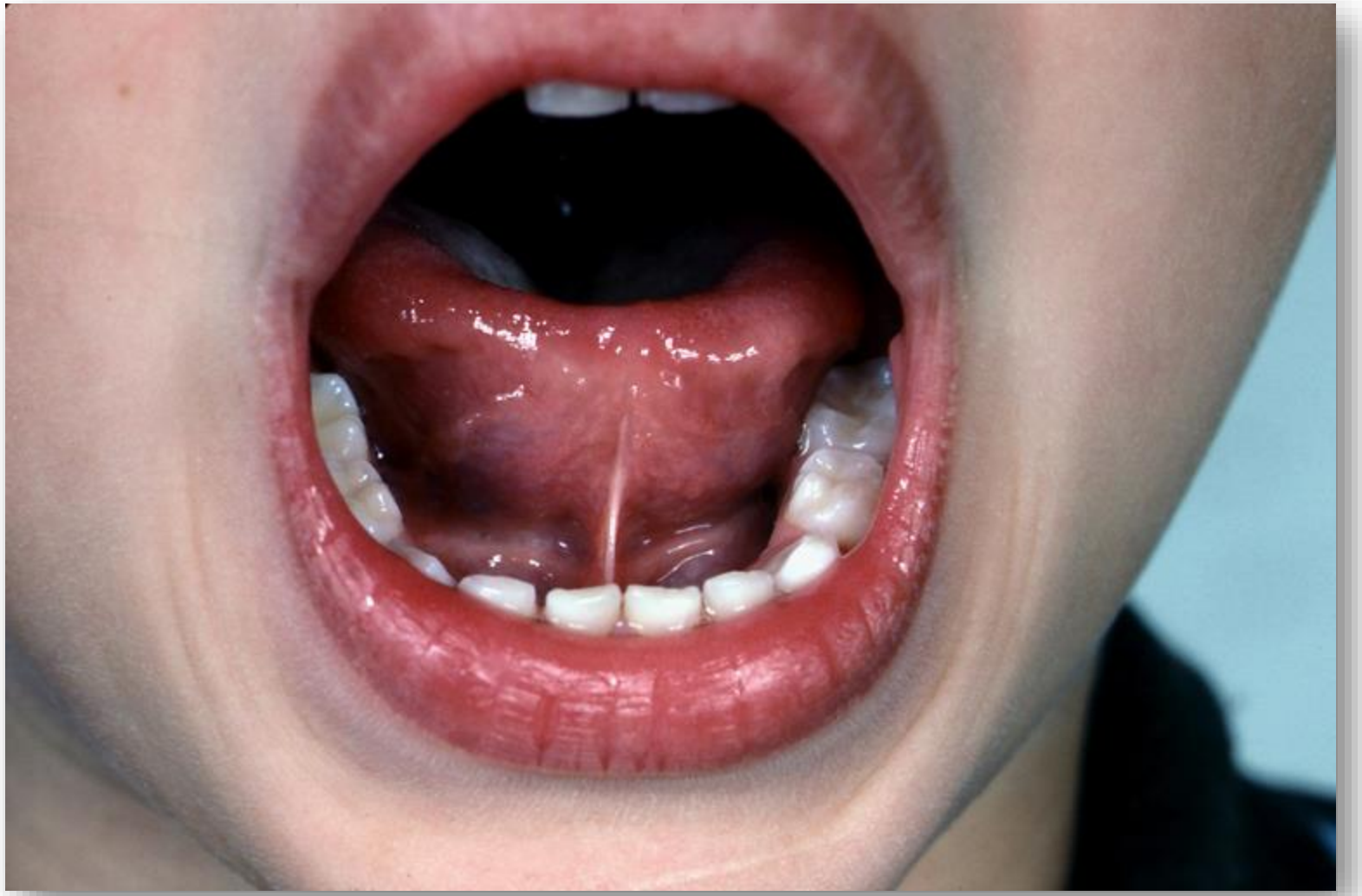

Scaphocephaly
Plagiocephaly
Brachycephaly
Trigonocephaly
Oxycephaly

Cleft lip

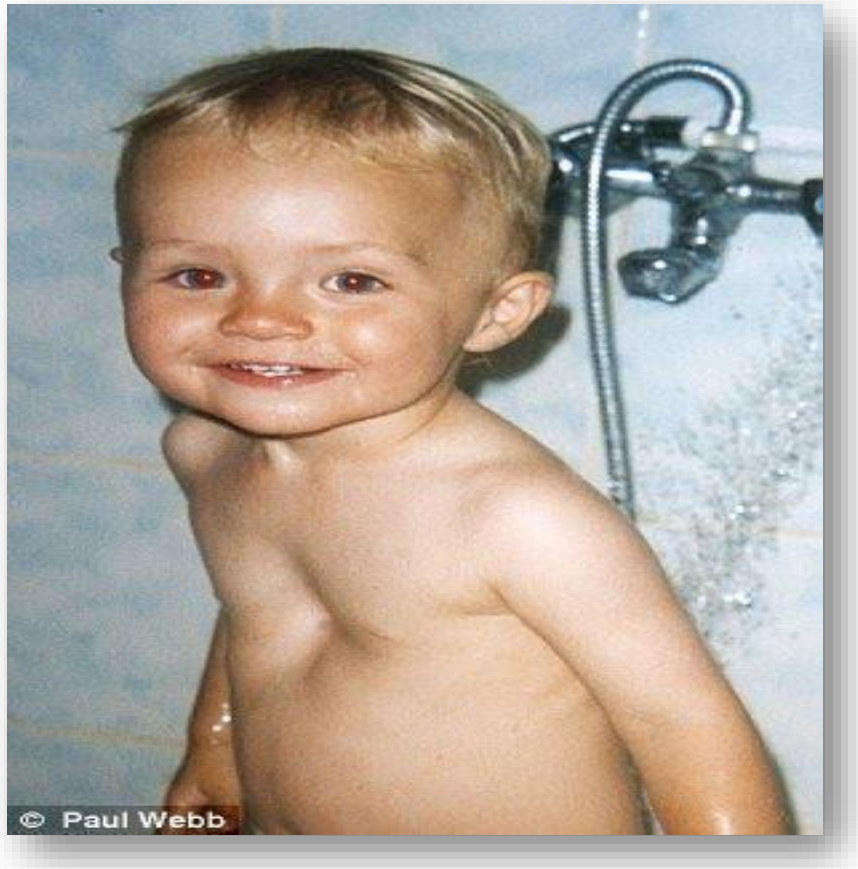
- **What's your Dx.?**
  - Unilateral complete cleft lip.
- **What are the risk factors to have this condition?**
  - Genetic (1st degree relatives, monozygotics "60% concordance").
  - Environmental (drugs "phenytoin, valproic acid, thalidomide",
  - Maternal alcohol & tobacco use, dioxins & other herbicides, & possibly high altitude).
- **What's the treatment? & when to be performed?**
  - Surgical closure; is usually done by 3 months of age.

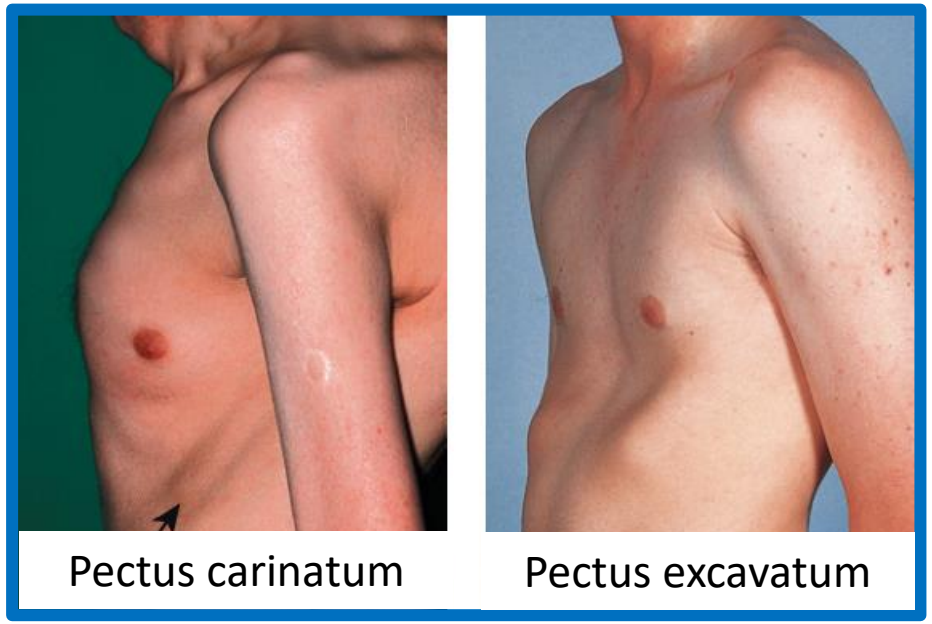
Ankyloglossia



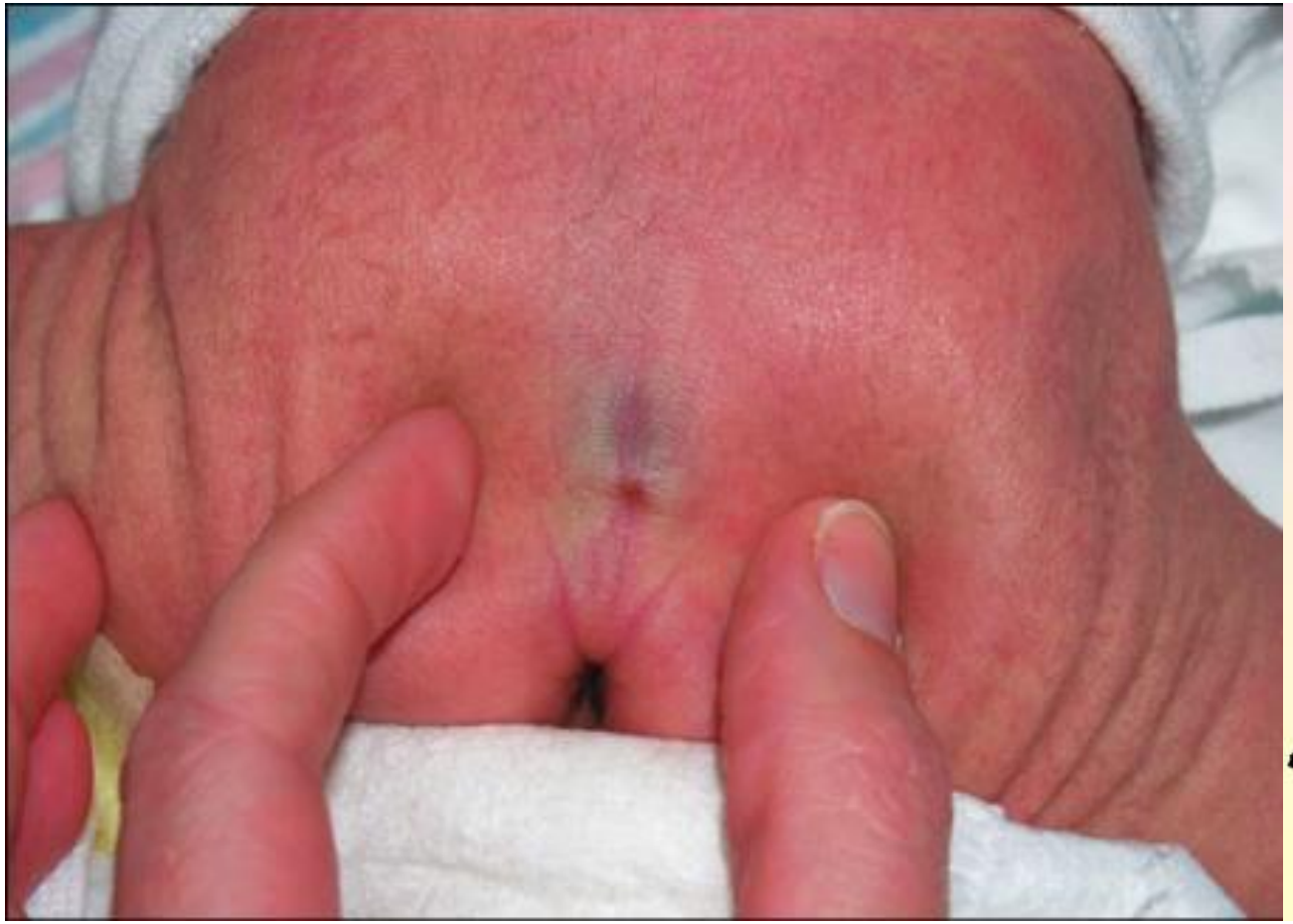
What is the name for this chest deformity ?

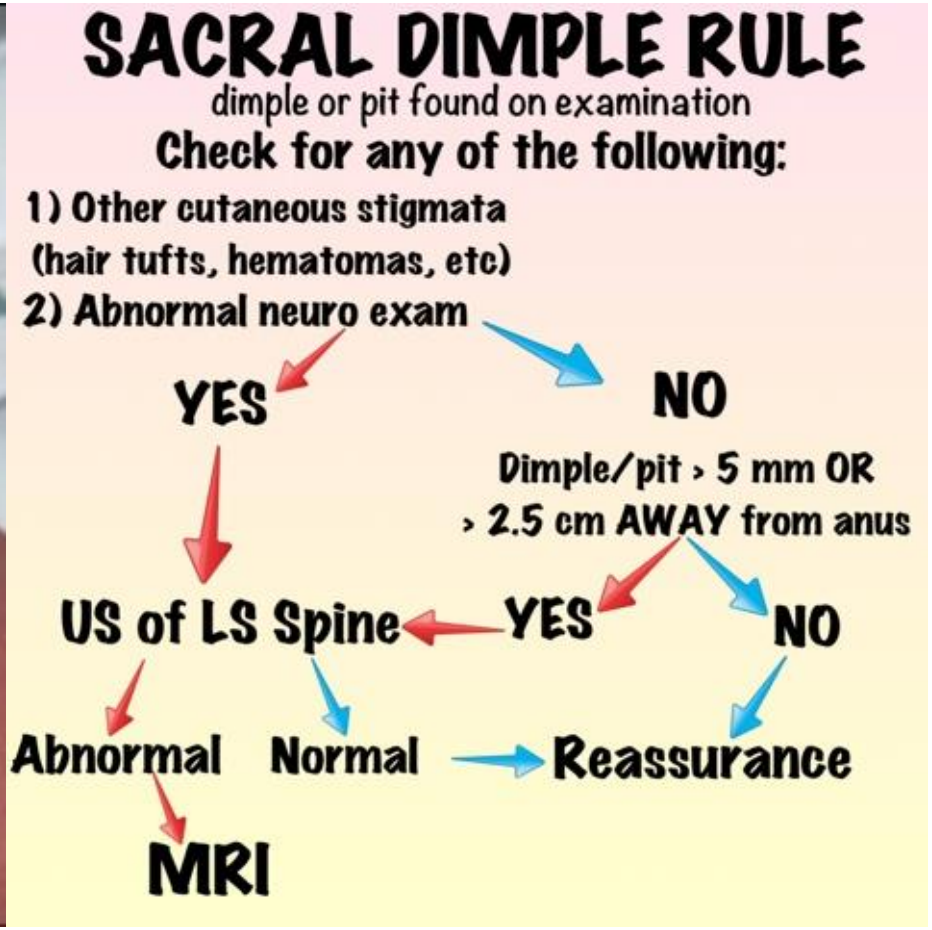
- Pectus excavatum





Sacral dimple

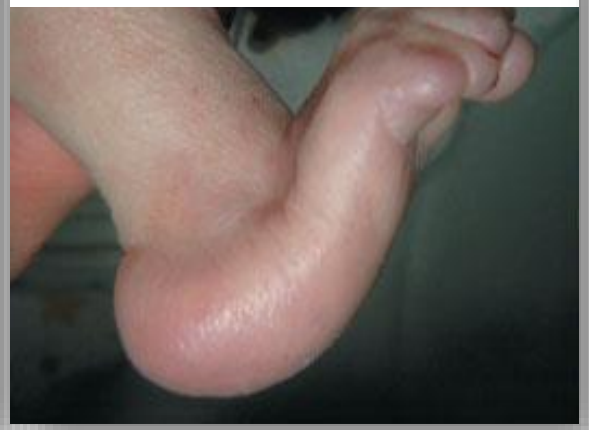


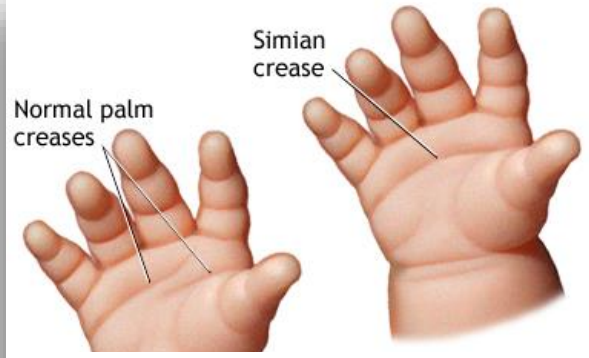


Dysmorphic features

Postaxial (ulnar); Common

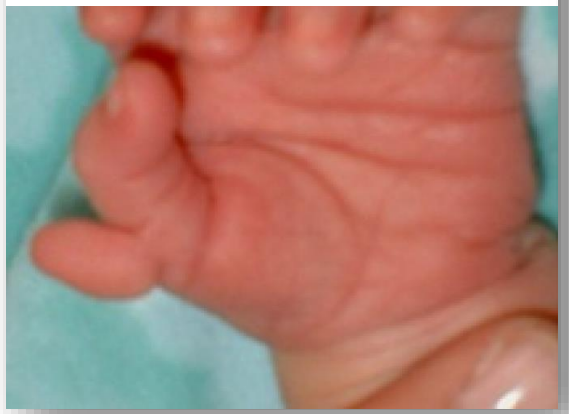
Rocker bottom feet



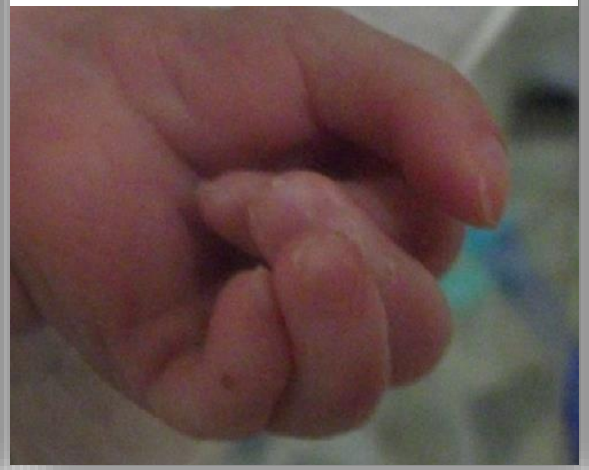


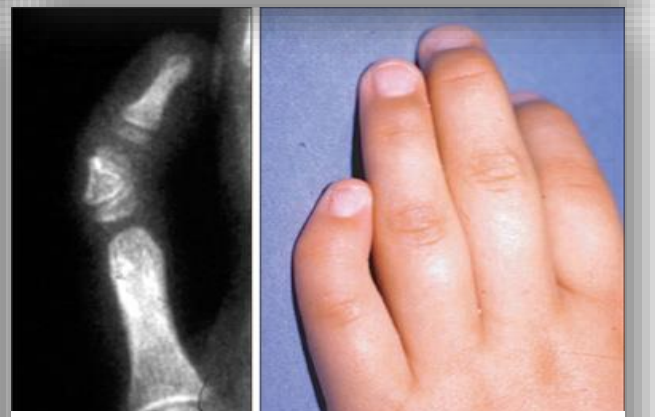
3-10% of normal population
May be associated with Down

Preaxial (radial); more associated with syndrome



Overriding fingers

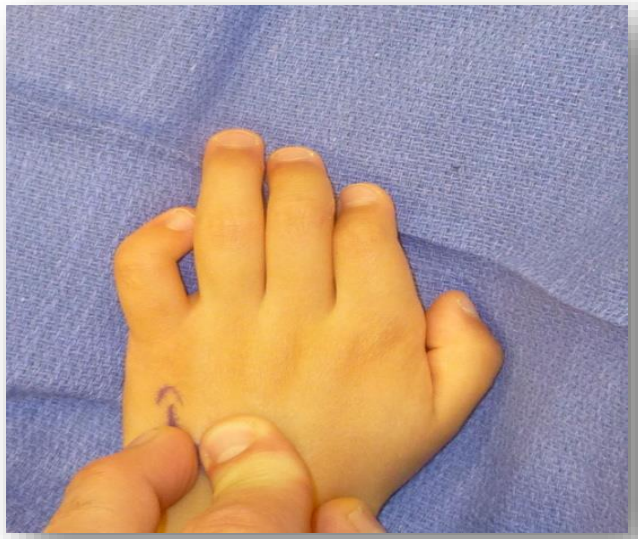
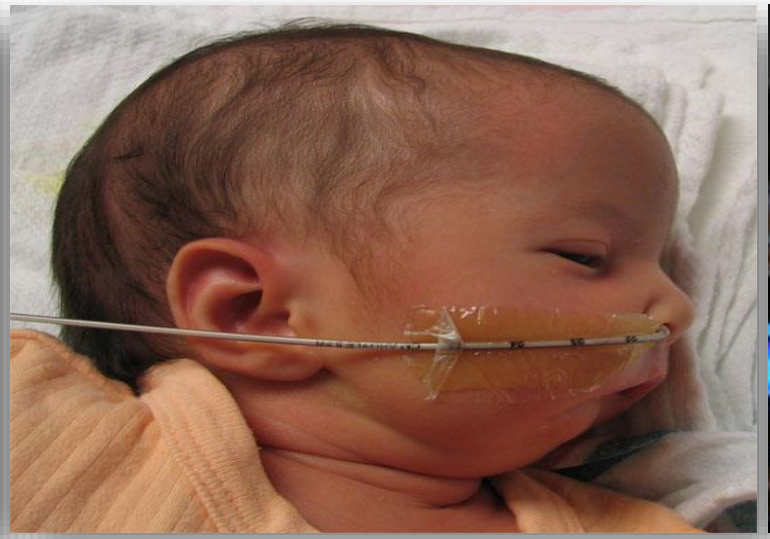




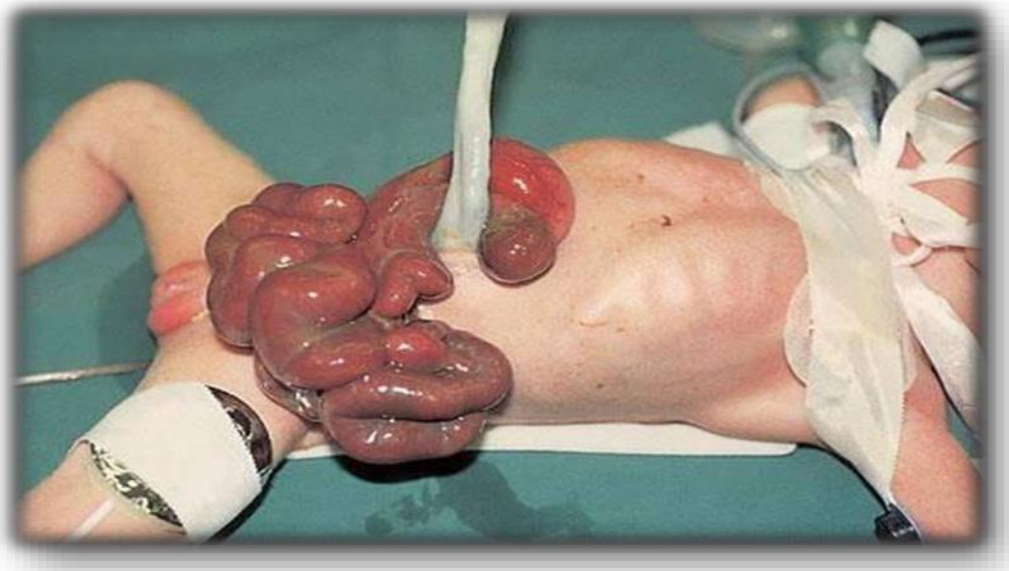
Clinodactyly: may be associated with Turner

Dysmorphic features

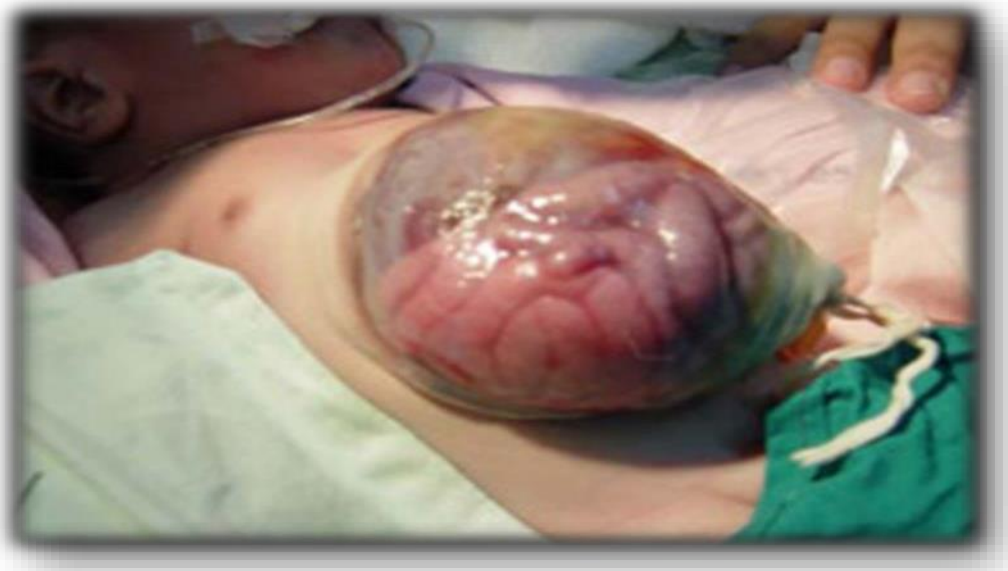
- **Write down 2 dysmorphic features you see**
  - Clinodactyly , Micrognathia, Macroglossia
- **What test you do to confirm dx ?**
  - Karyotyping


What is your Dx?



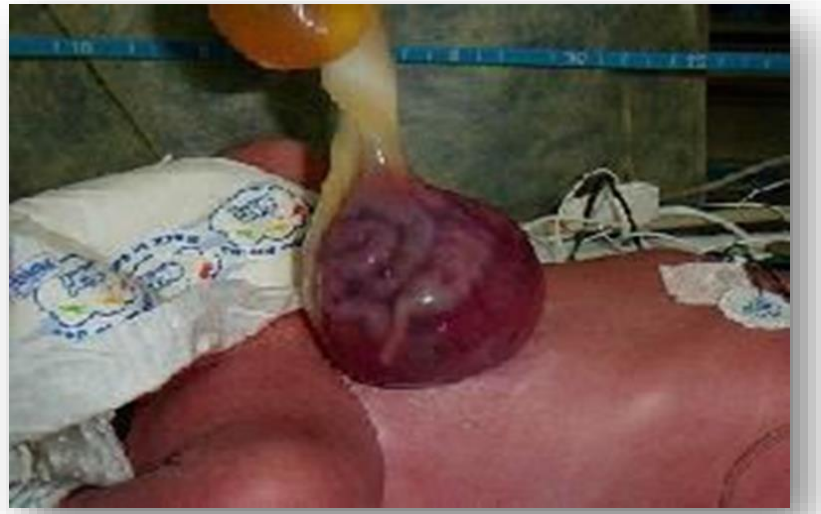
Gastroschisis



Omphalocele

Omphalocele

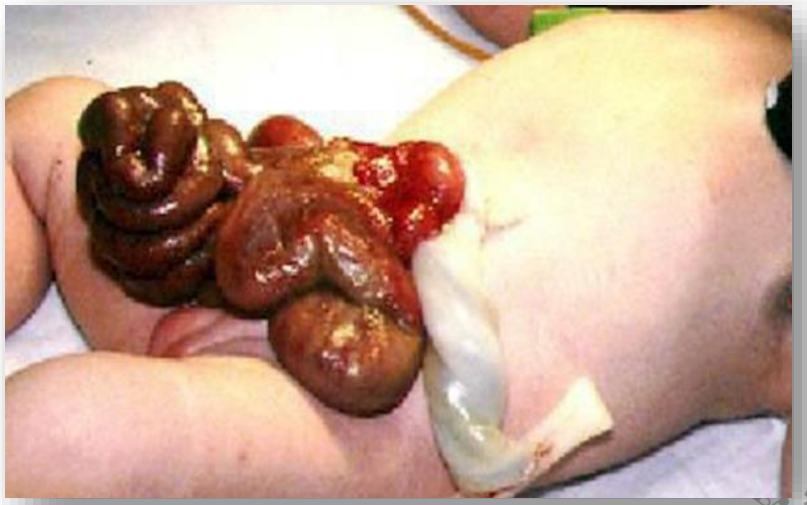
- **What's your Dx. ?**
  - Omphalocele
- **What other findings could be found in this patient ?**
  - Cardiac defects
  - Beckwith-Wiedemann syndrome (somatic overgrowth, hyper-insulinemic hypoglycemia, risk for Wilm's tumor)
  - Intestinal complications



Gastroschisis

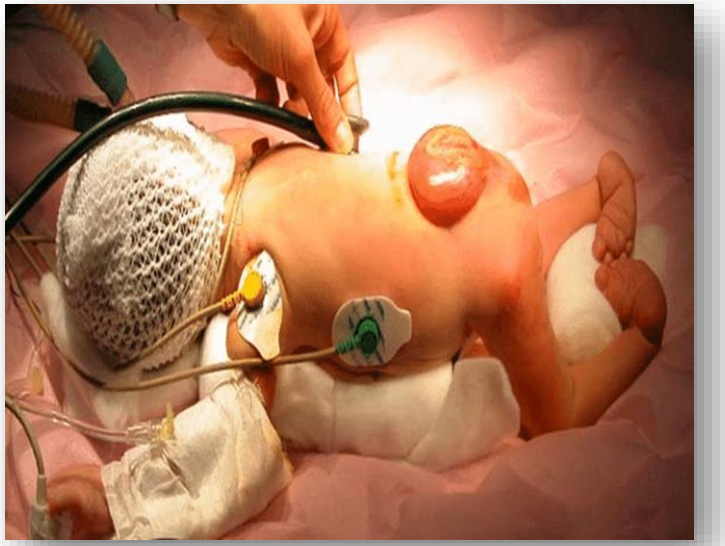
- **What's your Dx. ?**
  - Gastroschisis.
- **What's the importance of this condition ?**
  - Association with intestinal necrosis; although it's not associated with extra-intestinal anomalies, but segments of intestinal atresia are common.

- **Give 2 DDx.**
  - Gastroschisis, Omphalocele
- **What's the most important pre-op step of management ?**
  - Covering

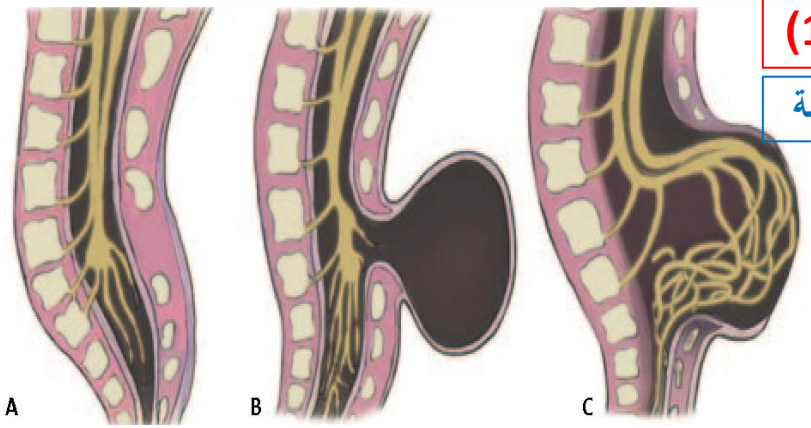


Spinal canal defects

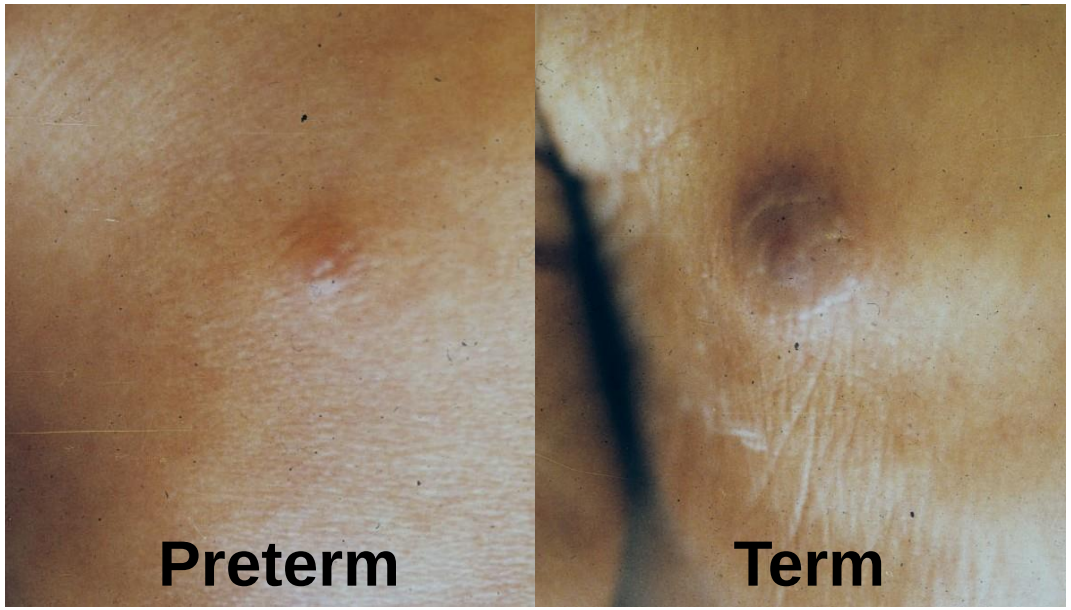
- **What is your diagnosis ?**
  - Myelomeningocele
- **Mention 1 drug that is given to prevent it**
  - Folic acid 400 micro
- **What expect about head circumference ?**
  - Microcephaly

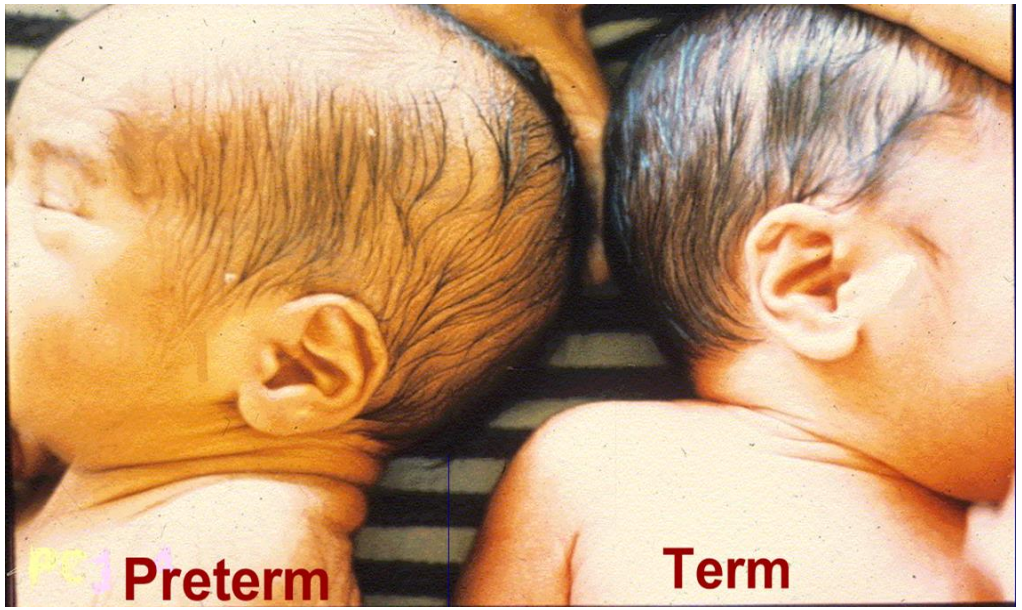


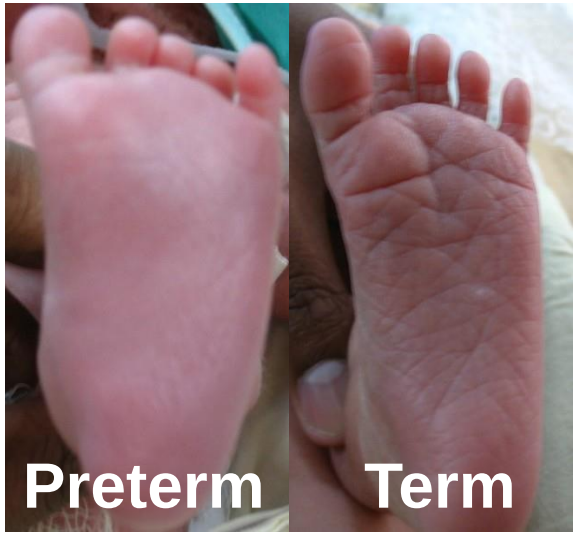
- **Name of the following**
  A. Spina bifida occulta
  B. Meningocele
  C. Myelomeningocele

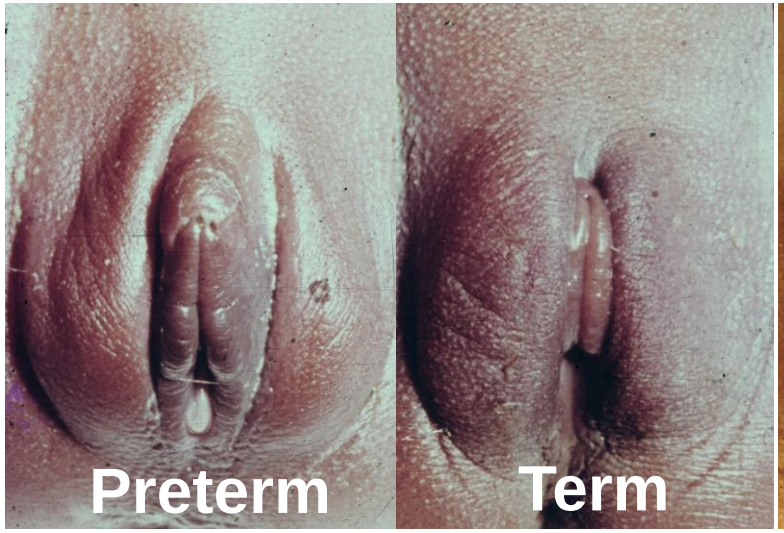


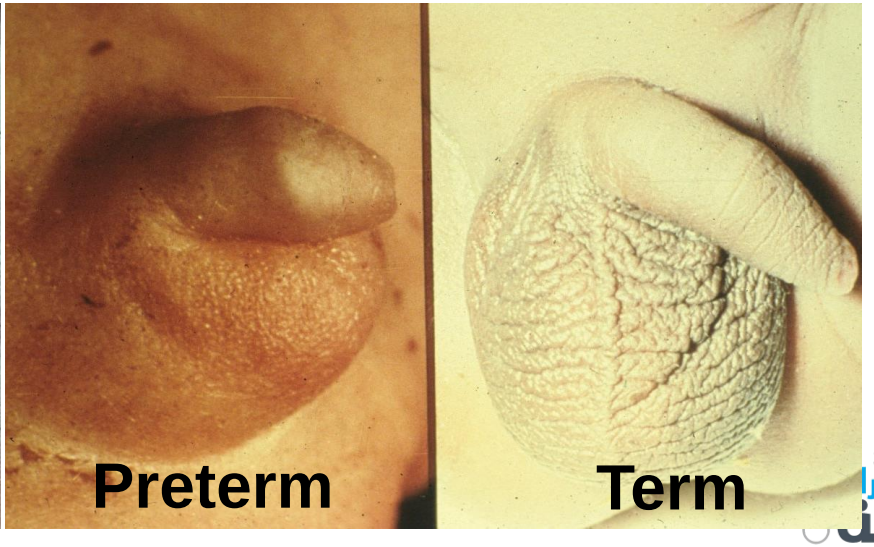
Preterm vs Term











Rickets

Medium-Yield

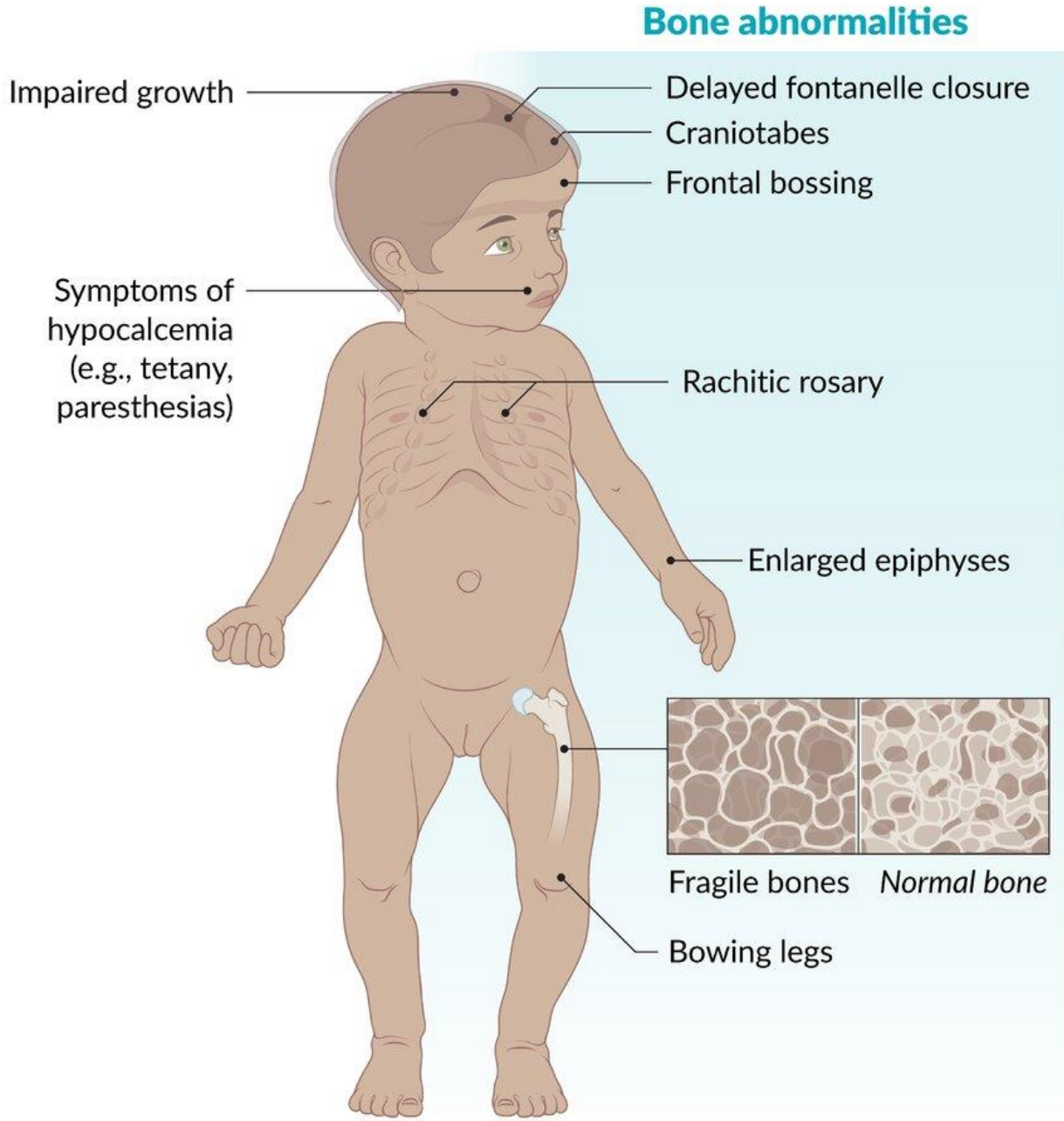
Rickets

Etiology
- Vitamin D deficiency due to:
  - Insufficient synthesis due to low UV radiation exposure (e.g., northern climates) and/or dark skin
  - Insufficient oral intake (e.g., in infants who are exclusively breastfed)
  - Malabsorption
  - Defective vitamin D metabolism

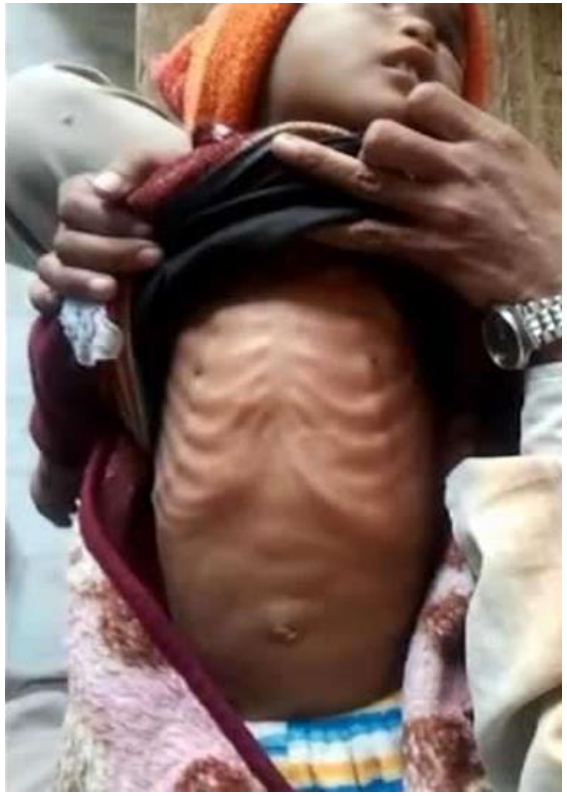
Pathophysiology
Vitamin D deficiency → defective mineralization of osteoid and growth plates

Diagnostics
↓ Vitamin D, ↓ serum Ca^{2+}, ↑ PTH, ↓ PO_4^{3-}, ↑ ALP

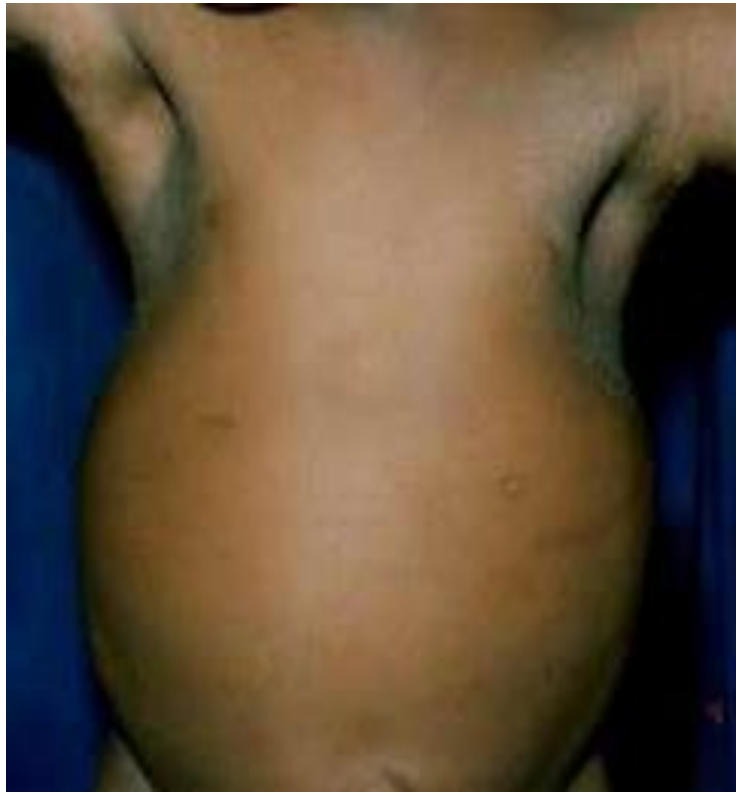
Treatment
- Dietary supplementation (vitamin D, calcium)
- Treatment of underlying cause



What are the names of the following deformities ?



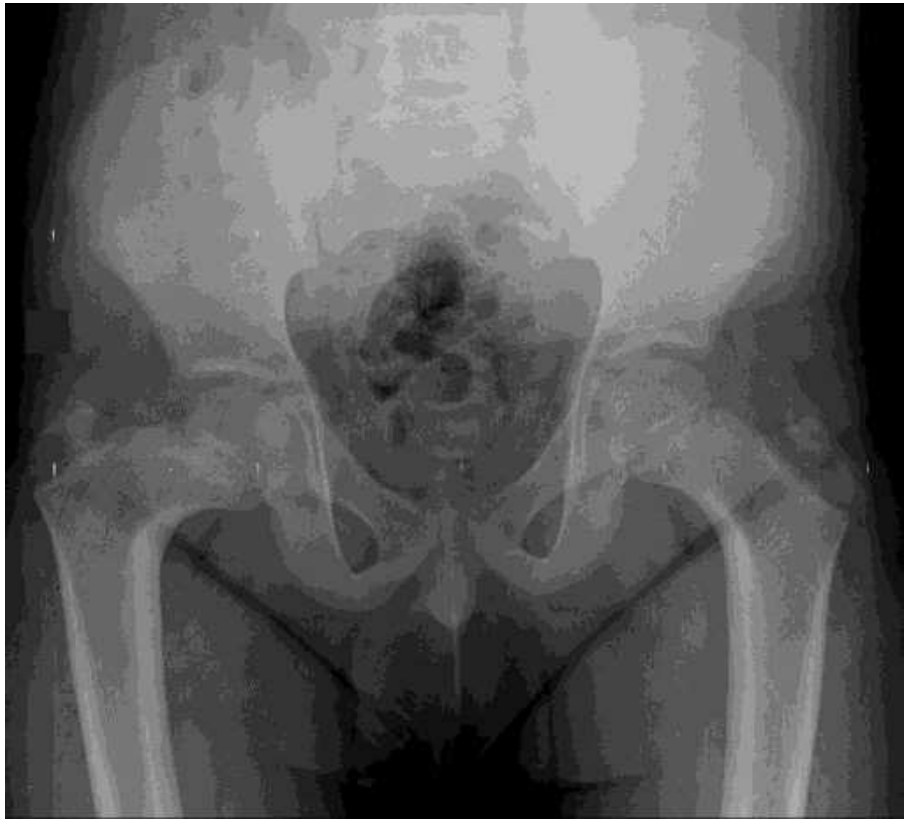
Rachitic rosary



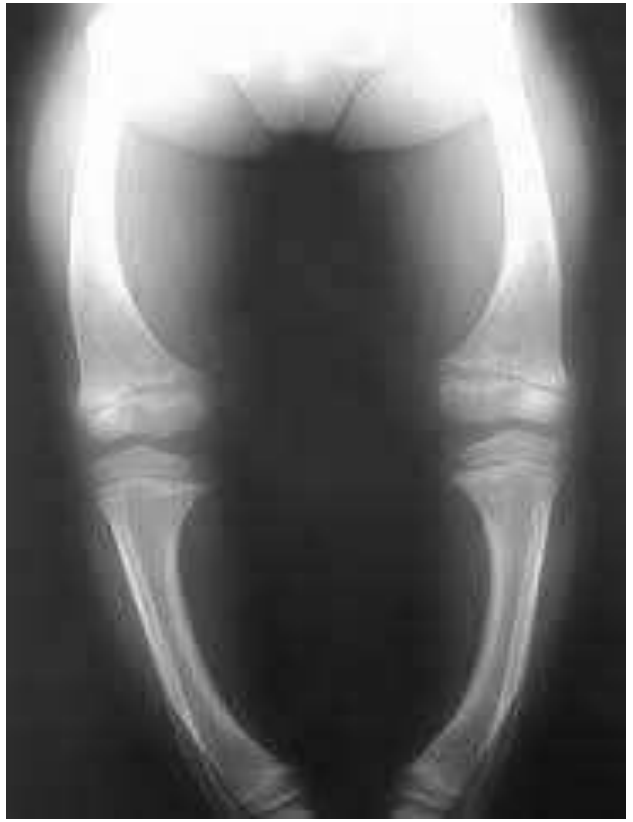
Harrison sulcus

Craniotabes

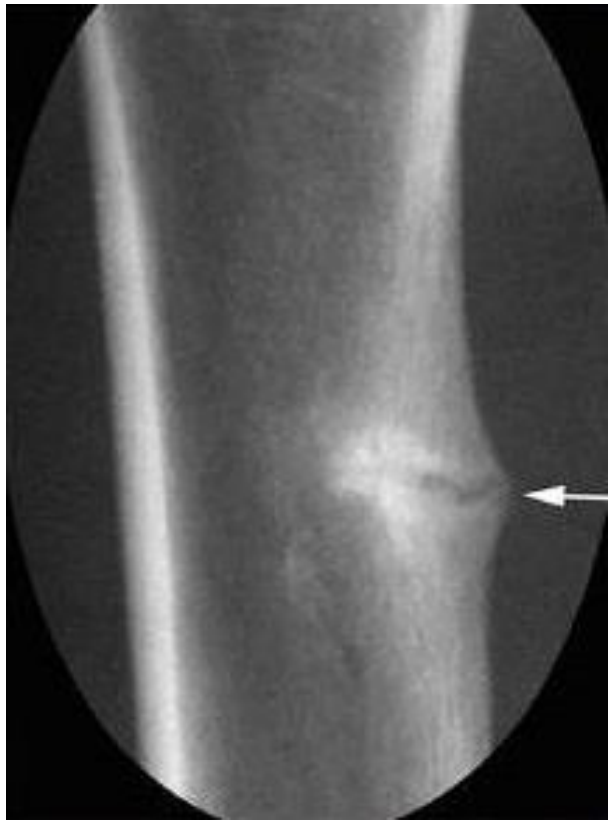
What are the names of the following deformities ?



Coxa vara



Genu vara



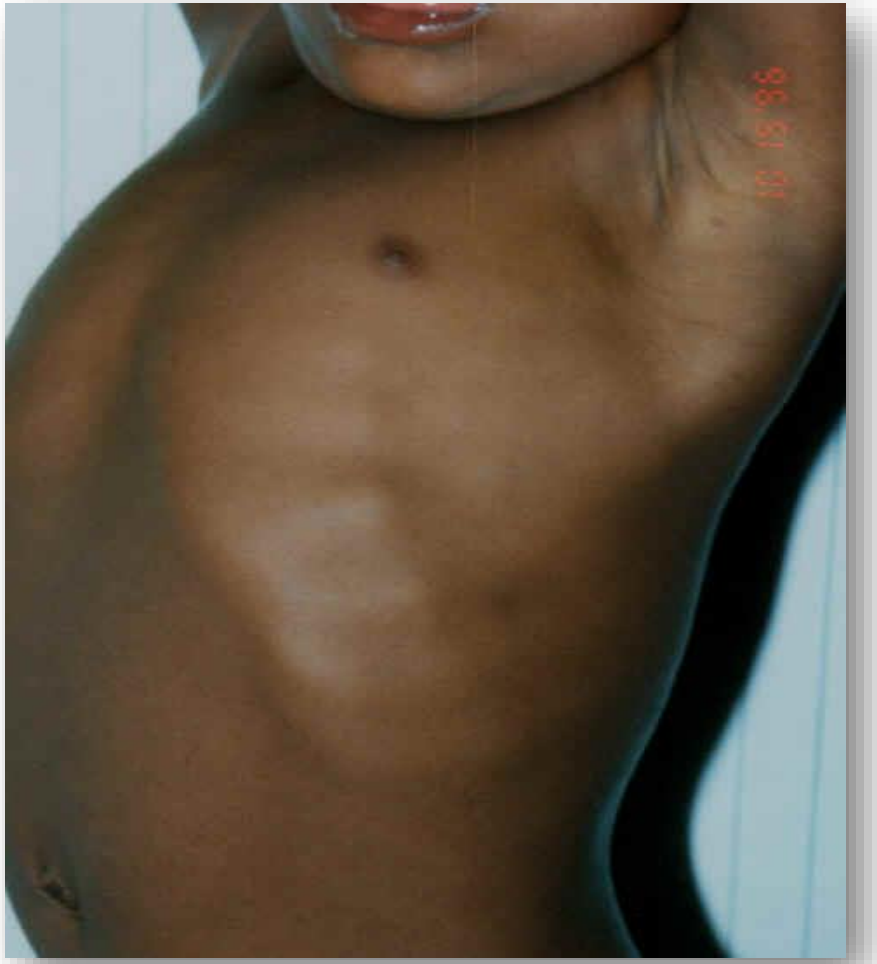
Looser fracture

Rachitic rosary sign

- **What is that sign ?**
  - Rachitic rosary sign
- **How to confirm your diagnosis ?**
  - Vit. D, PTH, Ca, PO_4, ALP

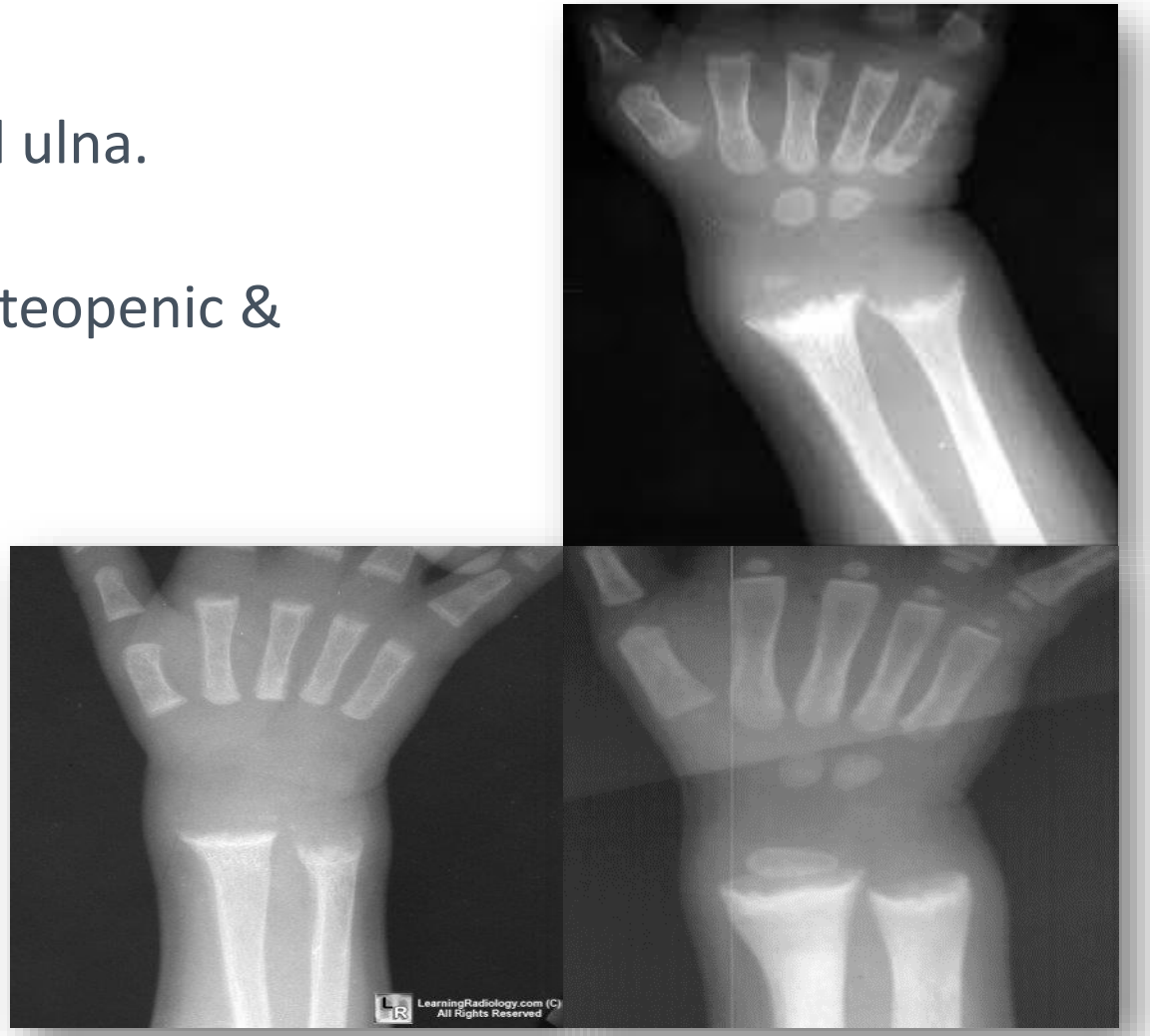
Rachitic rosary sign

- **What is that sign ?**
  - Rachitic rosary sign
- **Write other dysmorphic features ?**
  - Delayed fontanelle closure, Craniotabes, Frontal bossing, Harrison sulcus, Bowing
- **if Ca & Phosphate low, ALP high, what is the type of rickets ?**
  - Vitamin D dependent
- **if Ca low, Phosphate high, & ALP normal, what is the type of rickets ?**
  - Renal insufficiency



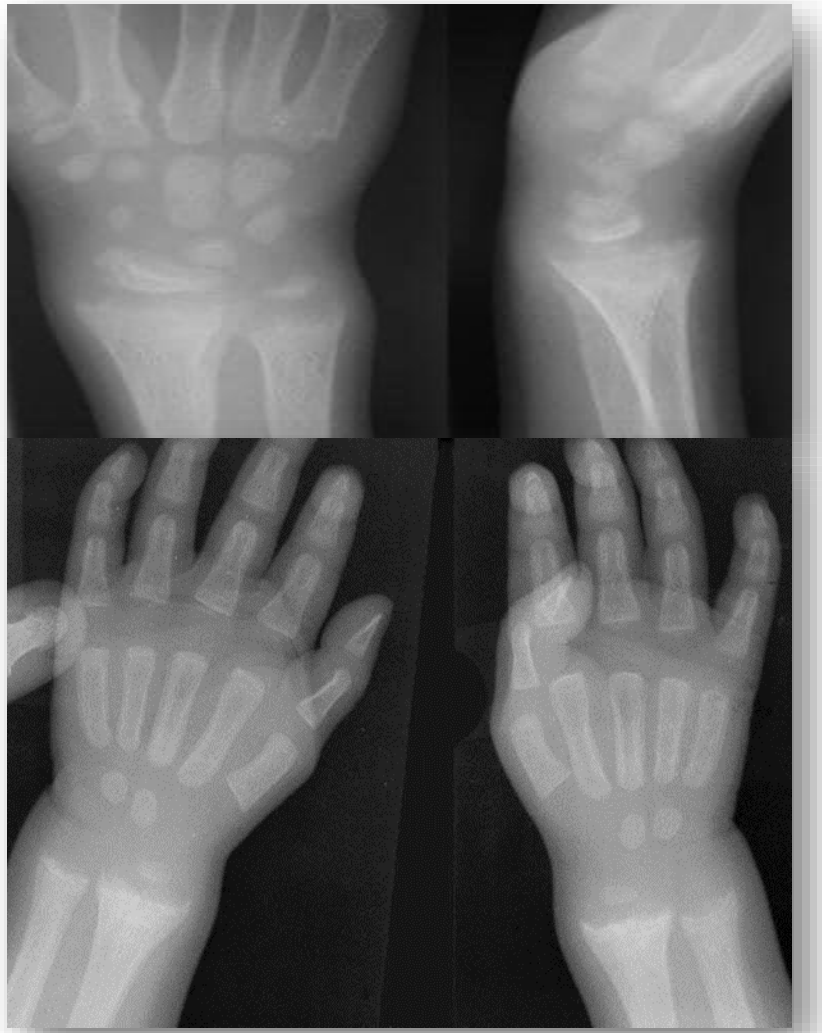
Cupping of distal head of radius and ulna

- **Findings is this photo ?**
  - Cupping of distal head of radius and ulna.
  - Widening of epiphyseal plate.
  - Shafts of the long bones become osteopenic & cortices become thin
- **What's your diagnosis ?**
  - Rickets
- **Investigations u need to do ?**
  - Vit. D, PTH, Ca, PO_4, ALP



Cupping of distal head of radius and ulna

- A patient who is exclusively breast fed presents with the following X-ray.



What is the radiological diagnosis?

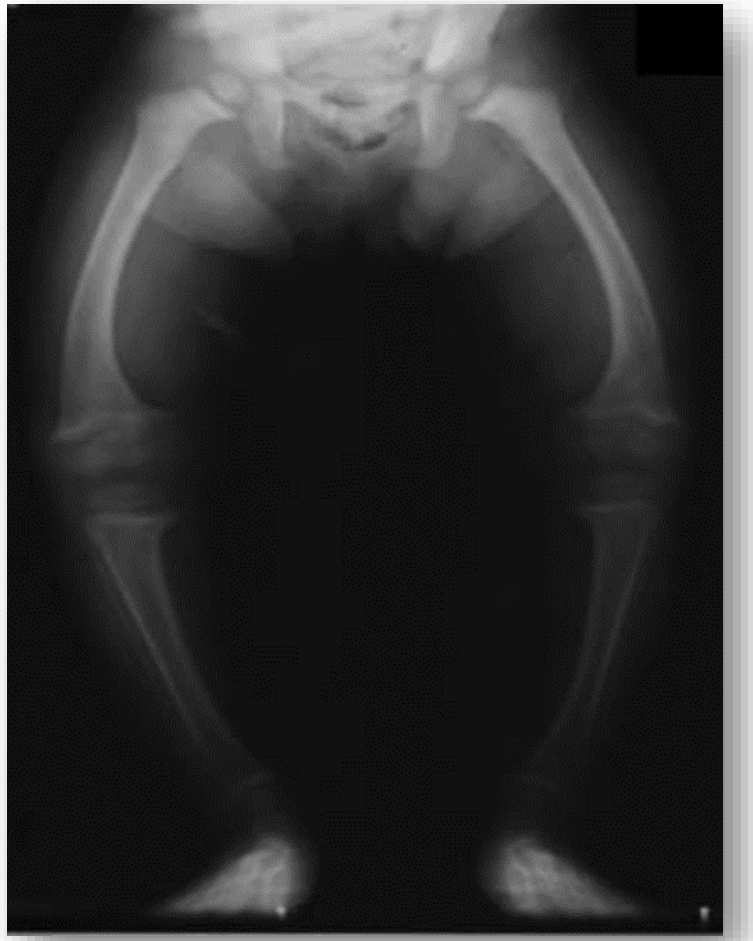
- Cupping of distal head of radius and ulna.
- Widening of epiphyseal plate.
- Shafts of the long bones become osteopenic & cortices become thin

Mention 2 abnormal labs?

- Low Ca+2
- Low Vitamin D
- Low Iron
- Low Fluoride

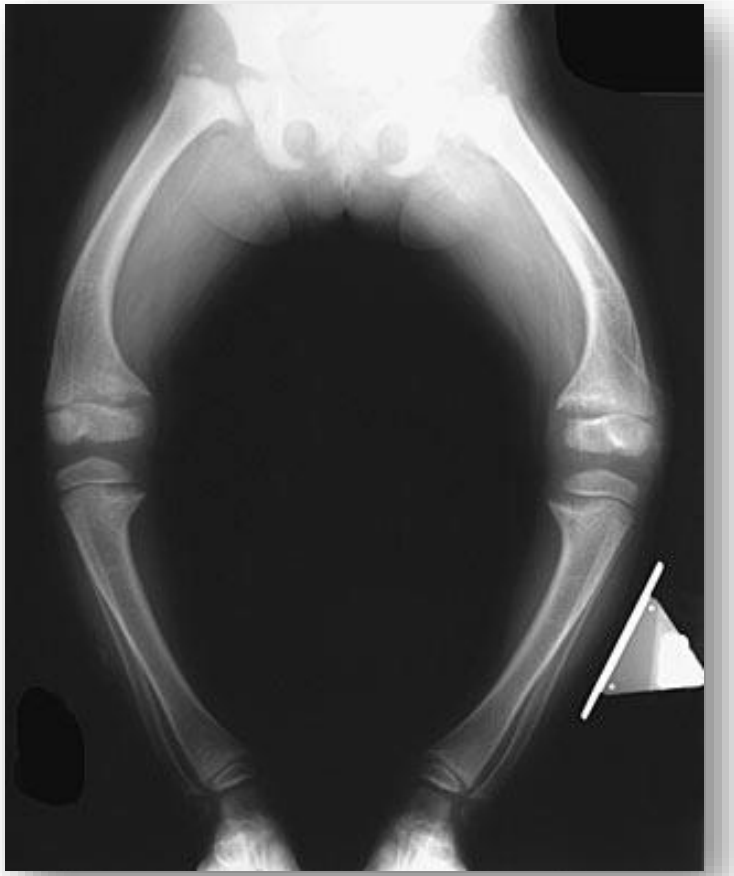
Bowing

- **What is your Dx ?**
  - Rickets
- **Give 2 abnormalities in the lower limbs**
  - Bowing
  - Valgus & varus deformity
  - Club foot
- **What is the test you will ask for ?**
  - Vit. D, PTH, Ca, PO_4, ALP
- **What is the treatment in this case ?**
  - Vitamin D with Ca+ supplement



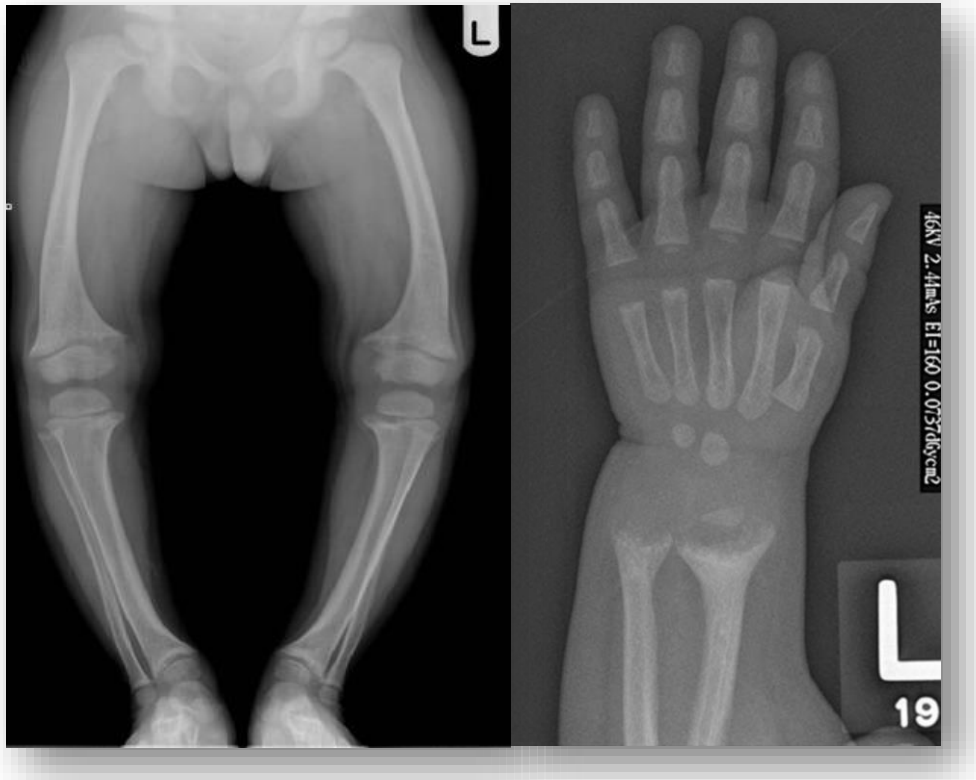
Bowing

- **What is your diagnosis ?**
  - Rickets
- **Daily maintenance dose of vit D**
  - 400-600 IU/day
- **If this patient has hypo PO4, aminoaciduria, and glycosuria ?** [سادسة]
  - Fanconi syndrome

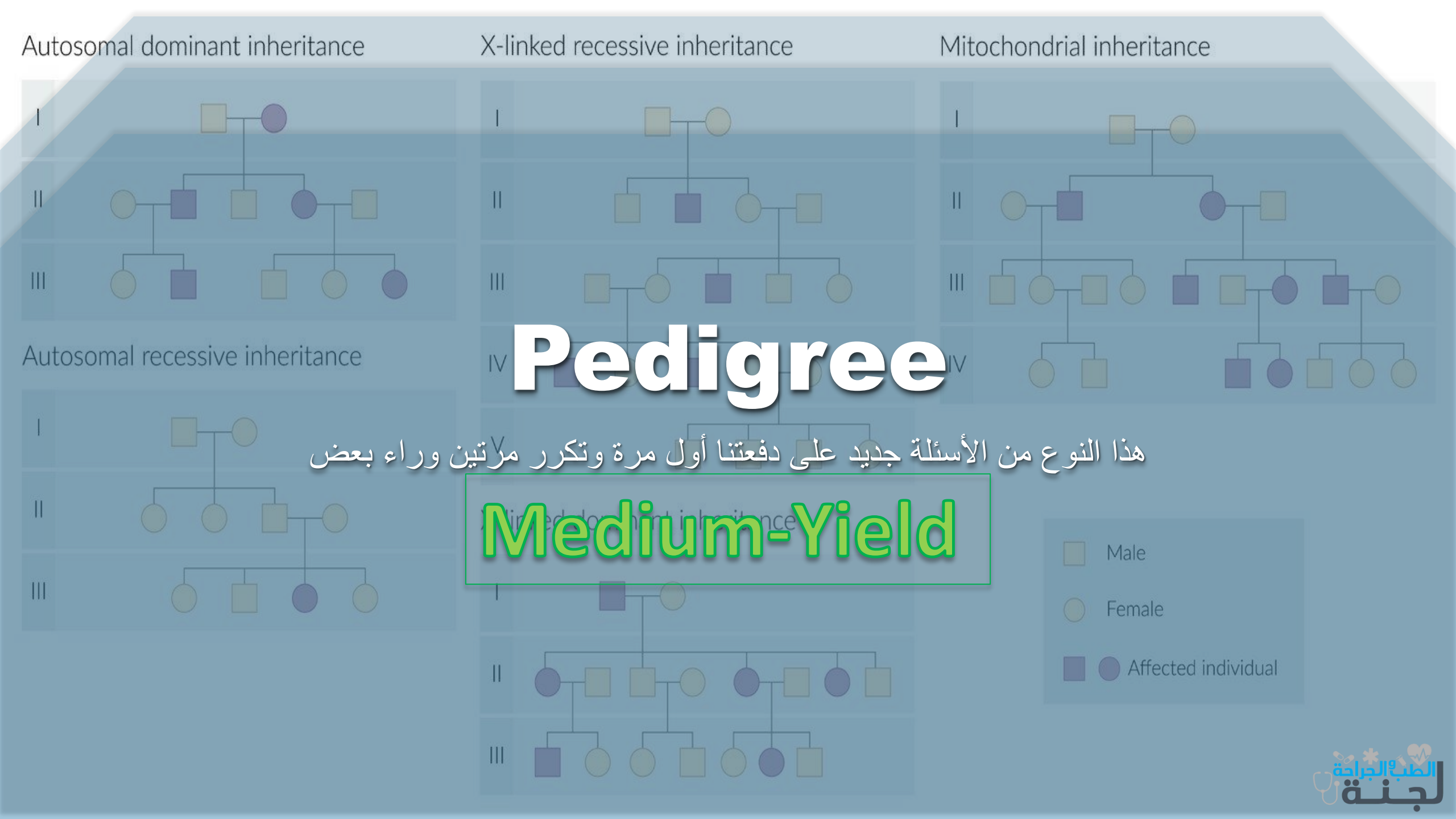


Rickets

- **What signs are seen in the pictures ?**
  - Splaying, Fraying, & Cupping
  - Genu varum / osteopenia
- **Mention 2 lab investigations you would like to order**
  - Serum vitamin D & serum calcium
- **What is the most likely diagnosis ?**
  - Rickets



Congenital & chromosomal anomalies



Pedigree

هذا النوع من الأسئلة جديد على دفعتنا أول مرة وتكرر مرتين وراء بعض

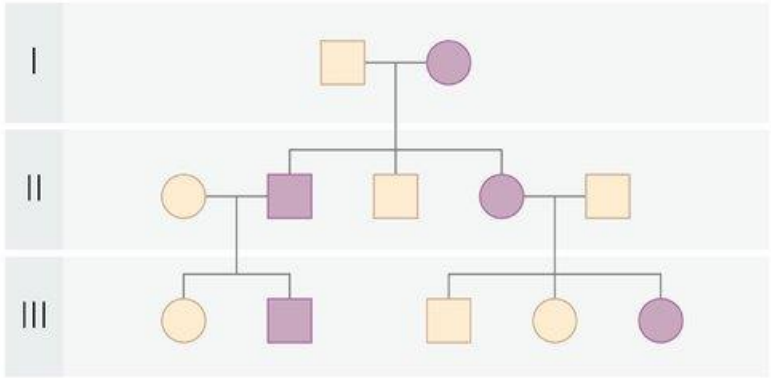
Medium-Yield

A basic methodological approach to pedigree analysis

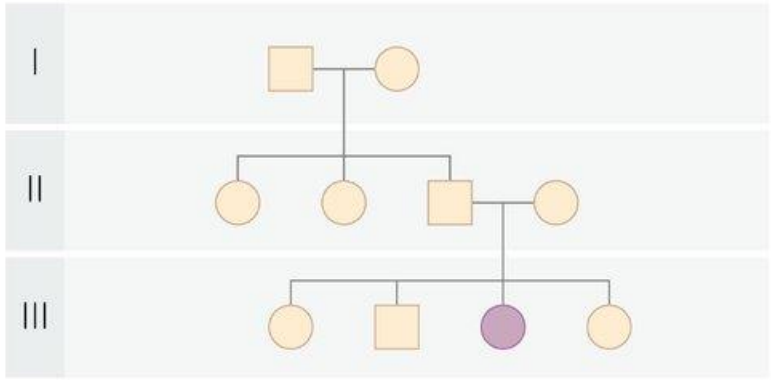
- Question 1: Does every affected family member have an affected parent?
  - Yes: dominant inheritance of the trait
  - No: recessive inheritance of the trait
- Question 2: Are the majority of affected family members male?
  - Yes: most likely X-linked recessive inheritance
  - No: Proceed to the following questions and subsequent answers.
    - Question 2a: Do all affected male family members have an affected mother?
    - Question 2b: Do all affected male family members have unaffected sons?
    - Question 2c: Do all affected male family members have affected daughters?
      - If Questions 2a, 2b, and 2c have all been answered in the affirmative, the disease most likely follows an X-linked dominant pattern of inheritance.
      - If Question 1 has been answered in the affirmative and either Question 2a, 2b, or 2c have been answered in the negative, the disorder is most likely autosomal dominant.
      - If Question 2 and Questions 2a, 2b, and 2c have all been answered in the negative, the disorder is most likely autosomal recessive.

إضافي

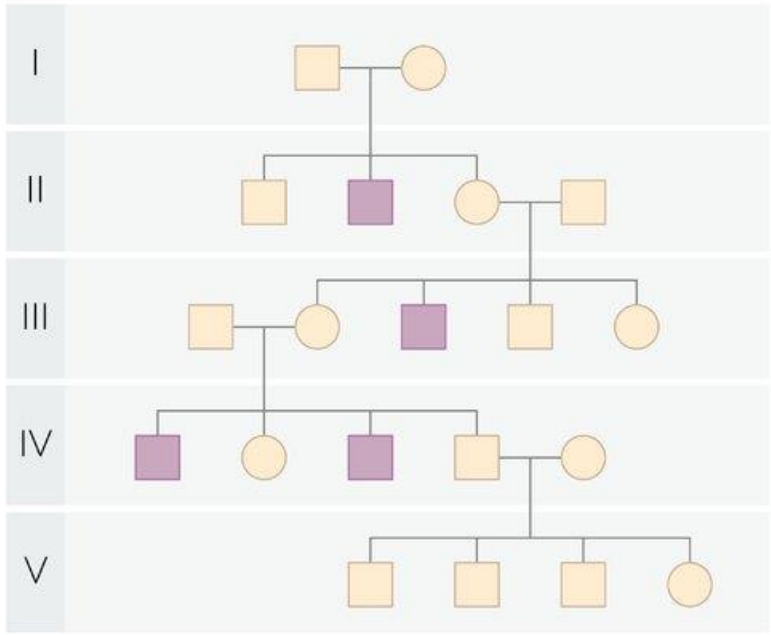
Autosomal dominant inheritance



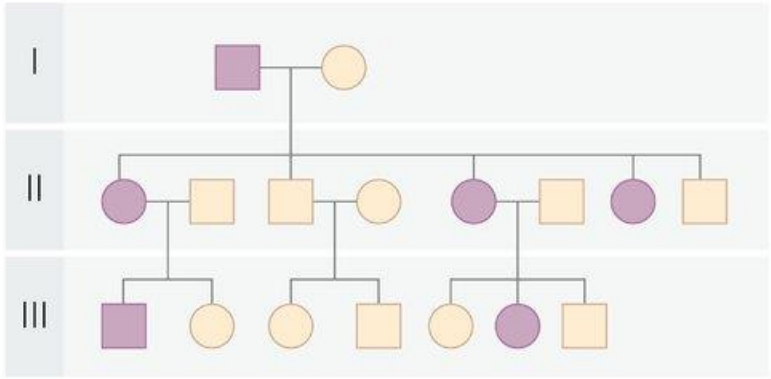
Autosomal recessive inheritance



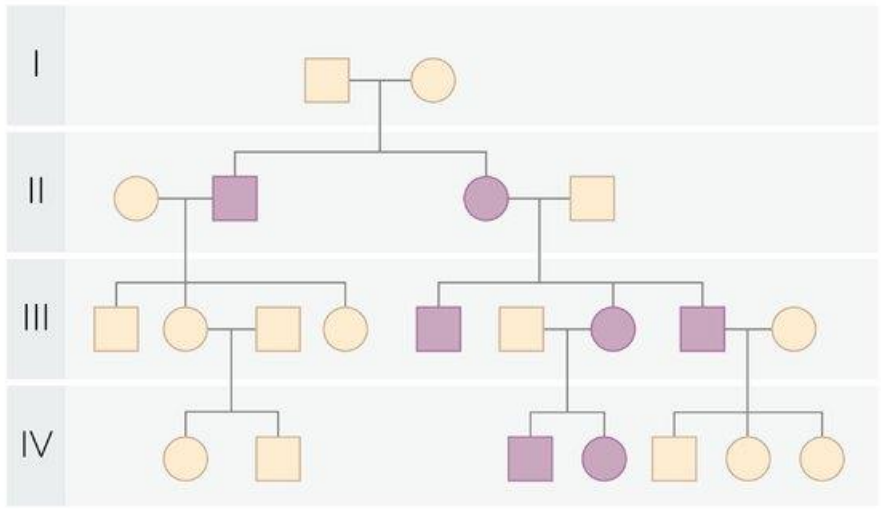
X-linked recessive inheritance

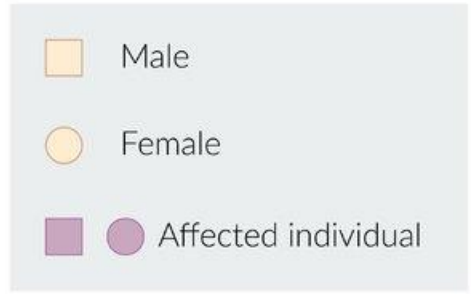


X-linked dominant inheritance



Mitochondrial inheritance





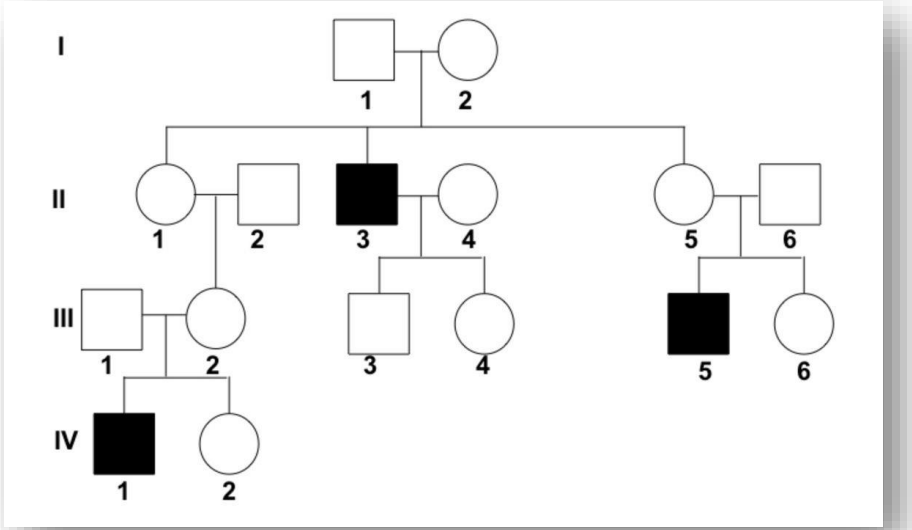
فيديوهات شرح أمبوس للموضوع

Part 1: Autosomal Inheritance Patterns

Part 2: Sex-linked Inheritance Patterns

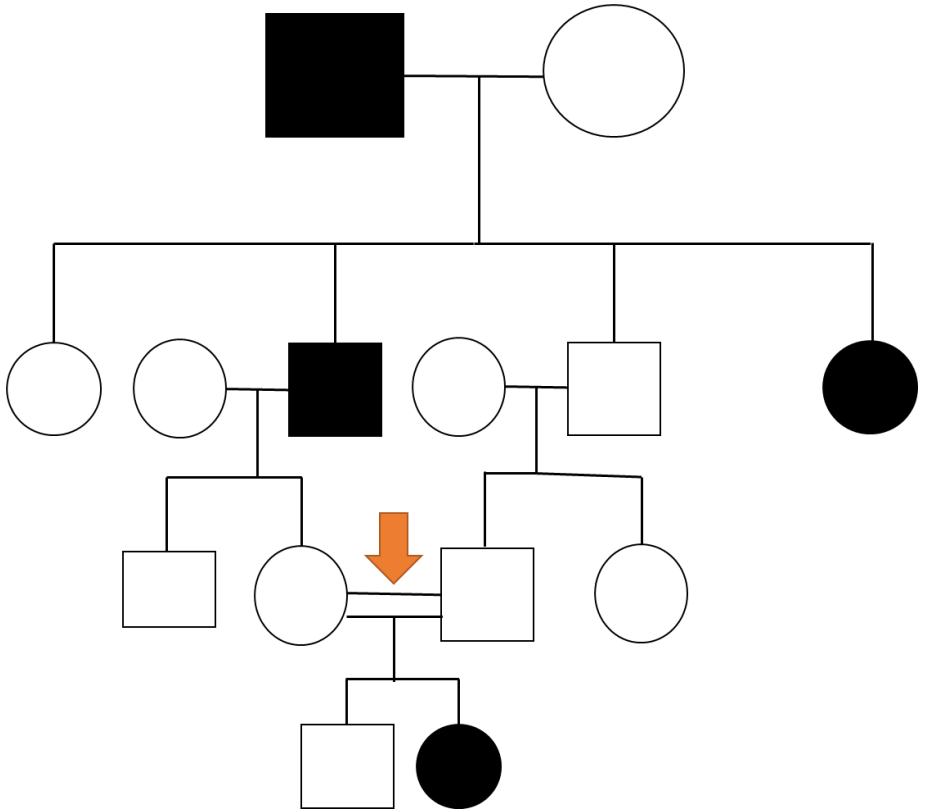
Pedigree

- **What is the type of inheritance ?**
  - X-linked recessive
- **Mention 2 diseases with this type of inheritance**
  - Wiskott-Aldrich syndrome
  - Hyper-IgM syndrome
  - Bruton agammaglobulinemia
  - chronic granulomatous disease
  - G6PD deficiency
  - Hemophilia A & B
  - Duchenne and Becker muscular dystrophy



Pedigree

- **What is the type of inheritance ?**
  - Autosomal recessive
- **Give two example for the type**
  - Cystic fibrosis
  - Hemophilia C
  - AR polycystic kidney disease
  - Friedreich ataxia
  - Hemochromatosis
  - Etc.
- **What does the arrowed double line indicate ?**
  - Consanguinity



إضافي

Examples of Autosomal disorders

Examples of Autosomal Dominant disorders	Examples of Autosomal Recessive disorders
• Achondroplasia • Autosomal dominant polycystic kidney disease (ADPKD) • Ehler-Danlos syndrome • Familial adenomatous polyposis • Familial hypercholesterolemia • Hereditary spherocytosis • Huntington disease • Li-Fraumeni syndrome • Marfan syndrome • Multiple endocrine neoplasias • Myotonic muscular dystrophy • Neurofibromatosis type 1 • Neurofibromatosis type 2 • Osler-Weber-Rendu syndrome • Osteogenesis imperfecta • Tuberous sclerosis • Von Hippel-Lindau disease	• Oculocutaneous albinism • ARPKD • Cystic fibrosis • Friedreich ataxia • Glycogen storage diseases • Hemochromatosis • Kartagener syndrome • Mucopolysaccharidoses (except Hunter syndrome, which is an X-linked recessive disorder) • Phenylketonuria • Sickle cell anemia • Sphingolipidoses (except Fabry disease, which is an X-linked recessive disorder) • Thalassemias • Wilson disease

Examples of X-linked disorders

Examples of X-linked Dominant disorders	Examples of X-linked Recessive disorders
• Alport syndrome • Fragile X syndrome • Hypophosphatemic rickets • Rett syndrome	• Becker muscular dystrophy • Bruton agammaglobulinemia • Color blindness • Duchenne muscular dystrophy • Fabry disease • G6PD deficiency • Hemophilia A • Hemophilia B • Hunter syndrome • Lesch-Nyhan syndrome • Ocular albinism • Ornithine transcarbamylase deficiency • Wiskott-Aldrich syndrome


Chromosomal anomalies
High-Yield

شرح

Down syndrome

Epidemiology
Incidence: ~ 1:700 live births

Etiology
Three complete copies of chromosome 21; due to meiotic nondisjunction in 95% of cases

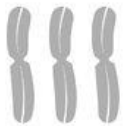
Karyotype
♀ : 47,XX,+21
♂ : 47,XY,+21

Complications
Due to organ malformations and immunodeficiency.
Increased risk of AML/ALL.
Early onset Alzheimer's disease

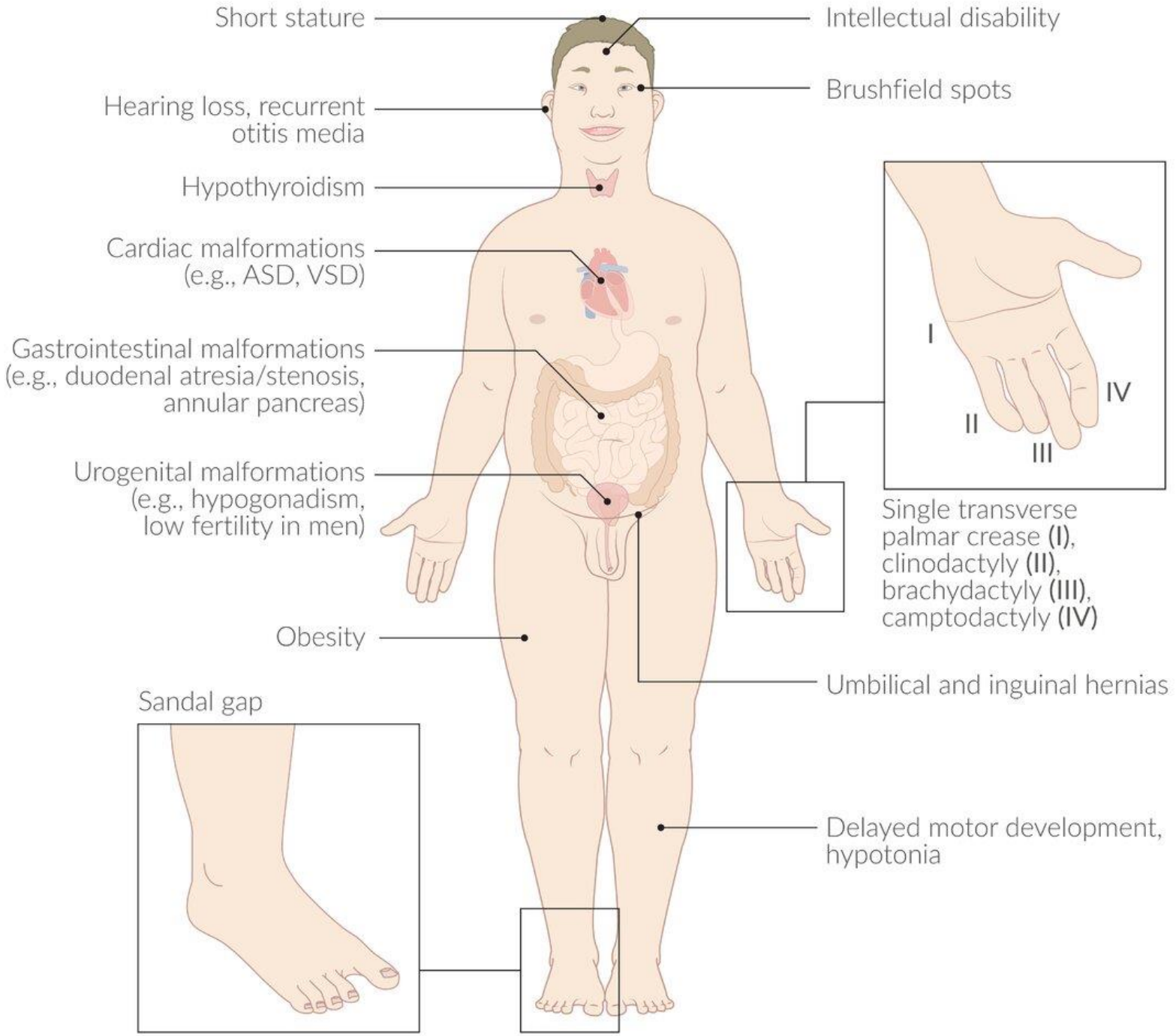
Important
Risk increases with maternal age

Life expectancy
~50 years

Karyotype

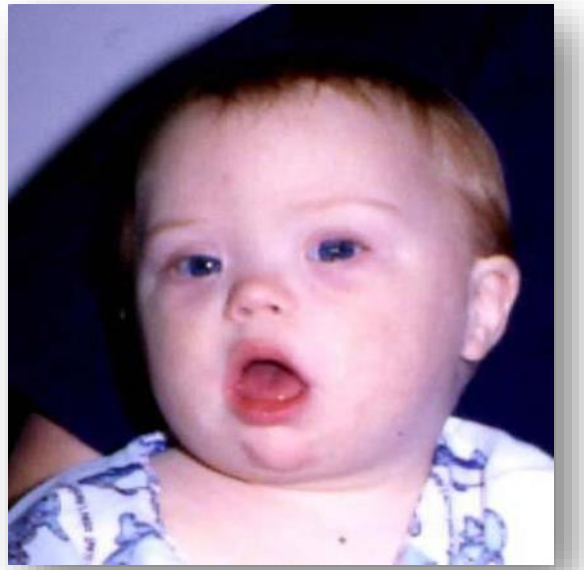


+21



Down syndrome

- **What's the most common cardiac anomaly associated with this problem?**
  - AV canal.
- **Mention 3 signs you can find at the hand.**
  - Clinodactyly, Caiman creases, Short fingers.
- **What syndrome is this?**
  - Down's Syndrome.
- **What is the chromosomal defect here?**
  - Trisomy 21



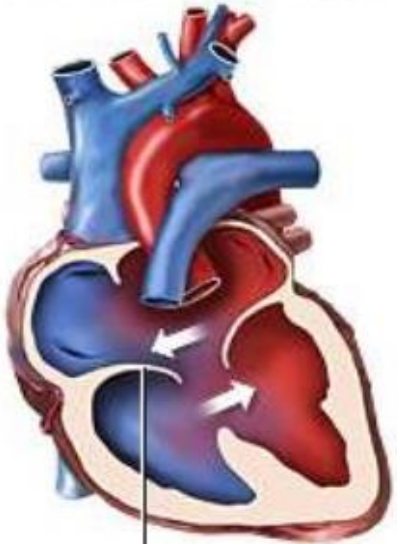
- **Name 2 congenital defects that are associated with this condition.**
  - AV canal (aka endocardial cushion defect), VSD, ASD, valvular disease, duodenal atresia, annular pancreas, & imperforate anus.

Down syndrome

- A child with down syndrome has this CXR, on examination there is a systolic murmur with no S3
- **What is your interpretation for this x-ray ?**
  - Cardiomegaly
- **What is the most common cause for it in this child ?**
  - Endocardial cushion defect

Down syndrome

- **What is the hematological disorder in this patient ?**
  - ALL , AML
- **Write one complication ?**
  - Heart failure
- **Name the heart defect you see**
  - Endocardial cushion defect



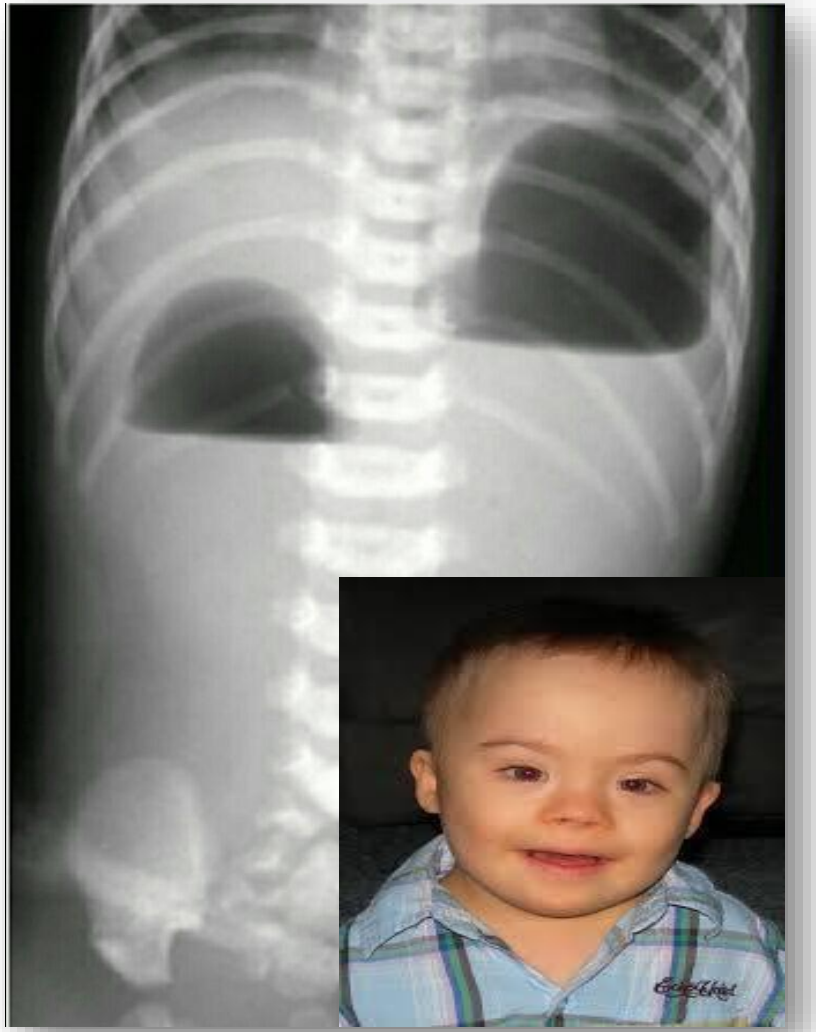
Down syndrome

- **Name 2 facial characters**
  - Hypertelorism, epicanthic fold, & micrognathia
- **What is the most common congenital anomaly in this patient?**
  - Endocardial Cushing defects (VSD > ASD > AV canal)
- **What is the most specific cardiac abnormality ?**
  - AV canal (endocardial cushion)
- **Name a gastrointestinal abnormality they have.**
  - Duodenal atresia, annular pancreas, Hirschsprung disease, imperforated anus

VSD is the most common cardiac anomaly even in down whereas AV canal is the most specific

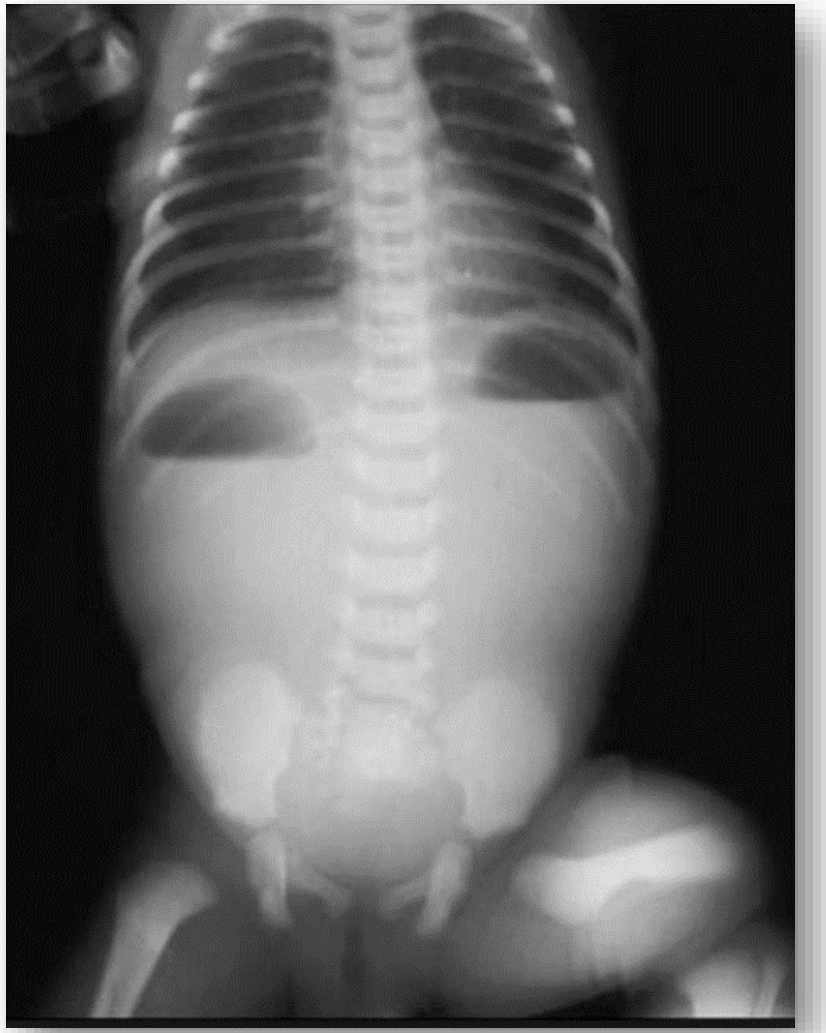
Down syndrome

- **What is this sign?**
  - Double-bubble sign.
- **What is the most probable diagnosis ?**
  - Duodenal atresia
- **What other signs do you expect on examination?**
  - Abdominal distension, Jaundice.
- **What would be the typical presentation?**
  - Bilious vomiting after the 1st feed.
- **Mention one risk factor for this condition**
  - Down syndrome



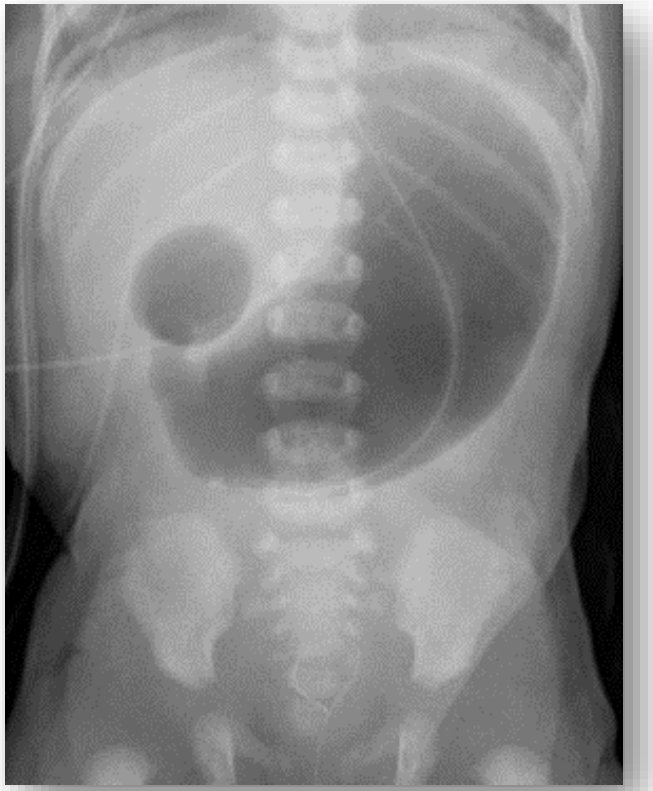
Double-bubble sign

- **What is the sign seen ?**
  - Double bubble sign
- **Give two possible GI diagnosis ?**
  - Duodenal atresia
  - Annular Pancreas



Duodenal atresia

- Newborn with bilious vomiting
- **What is the Cause of this X-ray finding ?**
  - Duodenal atresia
- **Mention 2 other causes of failure to pass meconium ?**
  - Hirschsprung's disease
  - imperforate anus

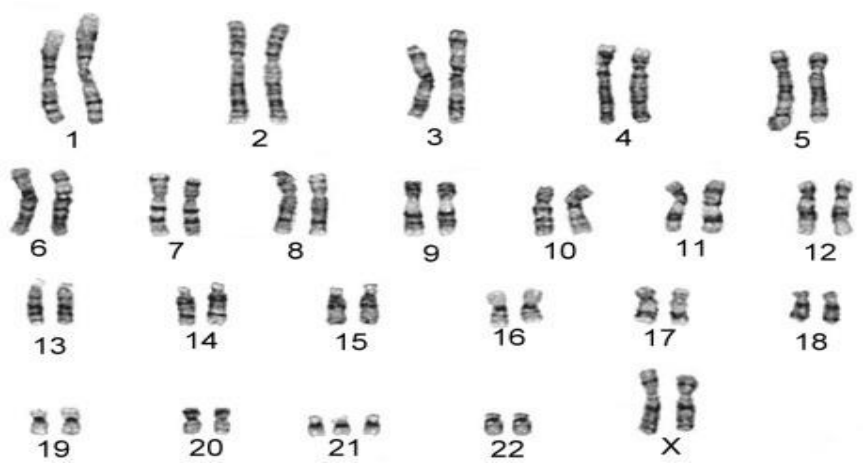


Down syndrome

- **What is the abnormality in the picture ?**
  - Brush-field spots
- **What condition is associated with this ?**
  - Down Syndrome

Down syndrome

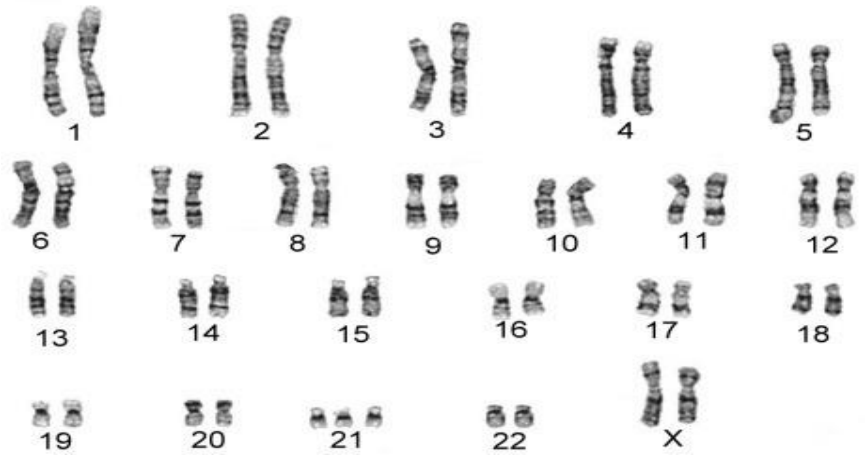
- **What is the diagnosis ?**
  - Trisomy 21 (Down syndrome)
- **What is the most likely cardiac defect ?**
  - Atrioventricular septal defect (AVSD)
- **Write two gastrointestinal anomaly would the patient have?**
  - Duodenal atresia / annular pancreas
- **What is the chromosomal abnormality during meiosis ?**
  - Non-disjunction of chromosome 21



Down syndrome

- **What is the most specific cardiac anomaly ?**
  - AV canal (endocardial cushion)
- **What is the lab test you want to check in 2nd trimester ?**
  - MS-AFP (decreased)
  - Estriol (decreased)
  - HCG (increased)
  - Inhibin A (increased)

- **Down etiologies:**
  - Non-disjunction ~92-95%
  - Robertsonian translocations ~4-5%
  - Mosaic Trisomy ~1-2%



شرح

Edwards syndrome

Epidemiology
Incidence: ~ 1:6.000
♀>♂

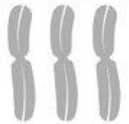
Etiology
Presence of an extra chromosome 18

Karyotype
♀ : 47,XX+18
♂ : 47,XY+18

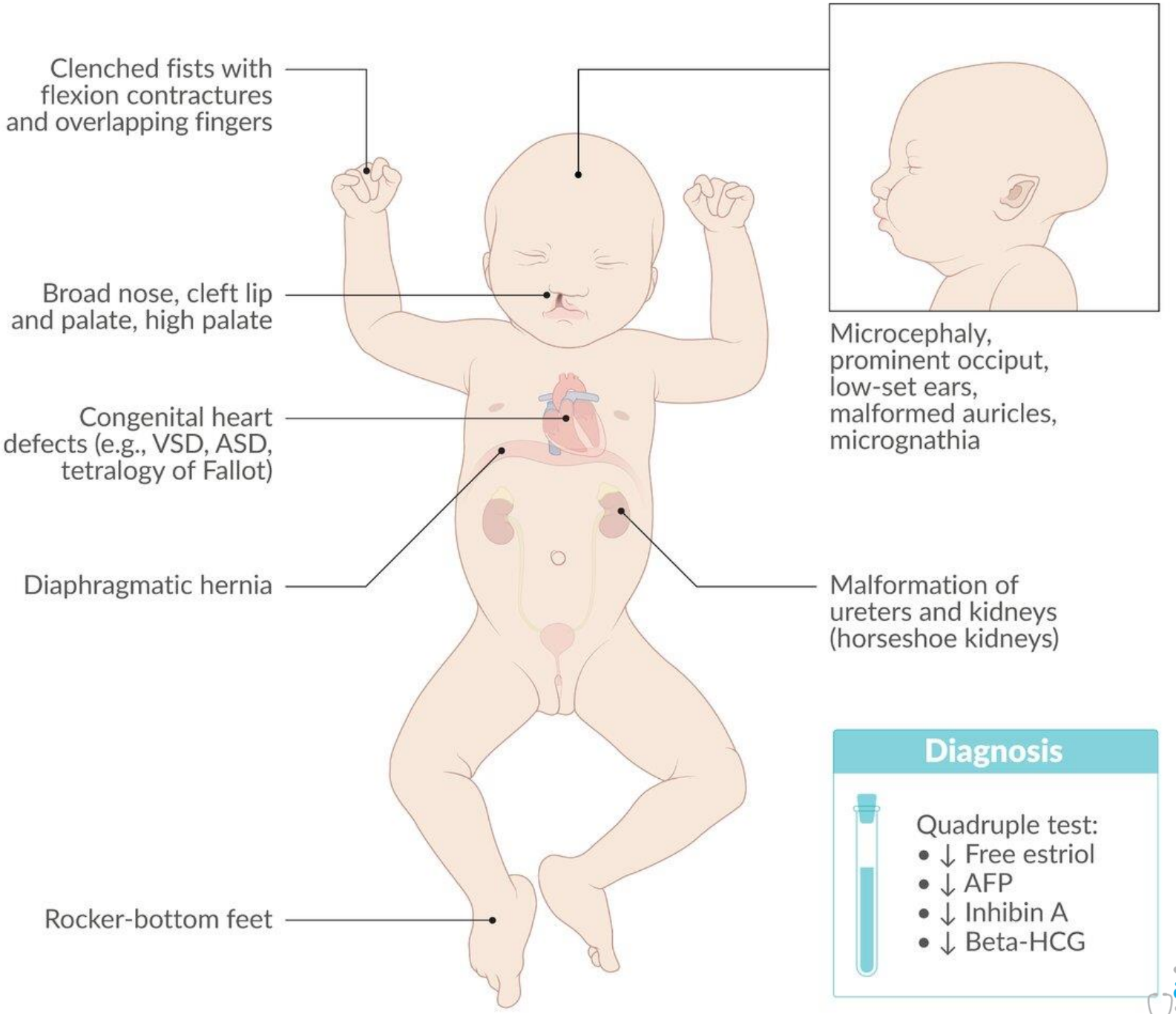
Important
Second most common trisomy after Down syndrome (trisomy 21); risk increases with maternal age

Life expectancy
Only 5–10% survive past 12 months of age

Karyotype



+18

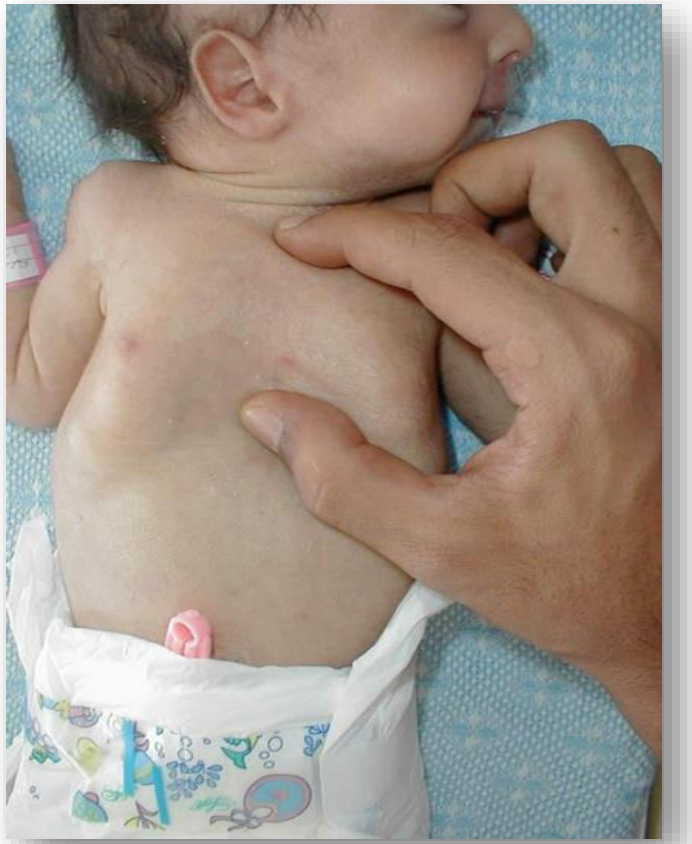


Diagnosis

Quadruple test:
- ↓ Free estriol
- ↓ AFP
- ↓ Inhibin A
- ↓ Beta-HCG

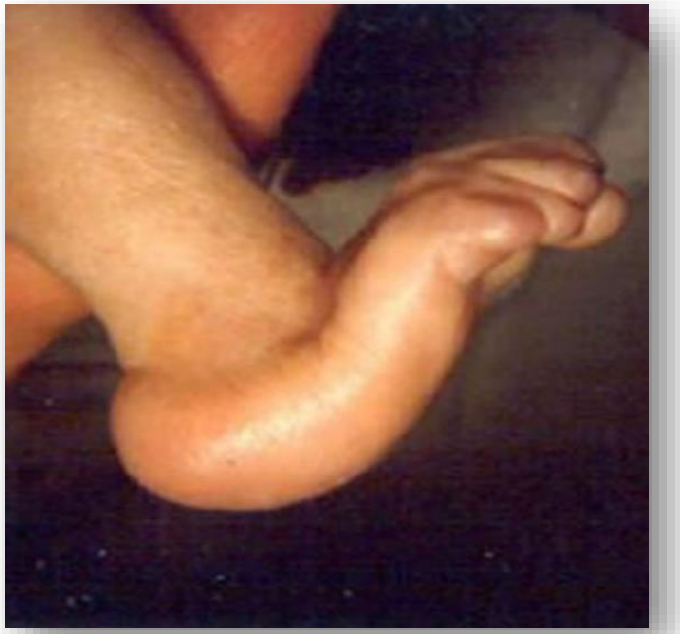
Edwards syndrome

- **What's the name of this sign ?**
  - Short sternum
- **What's the most likely Diagnosis ?**
  - Edwards syndrome (Trisomy 18)
- **Give other 2 physical findings related to this disorder (not seen in the image)**
  - Microcephaly
  - Ocular hypertelorism
  - Low-set, malformed ears



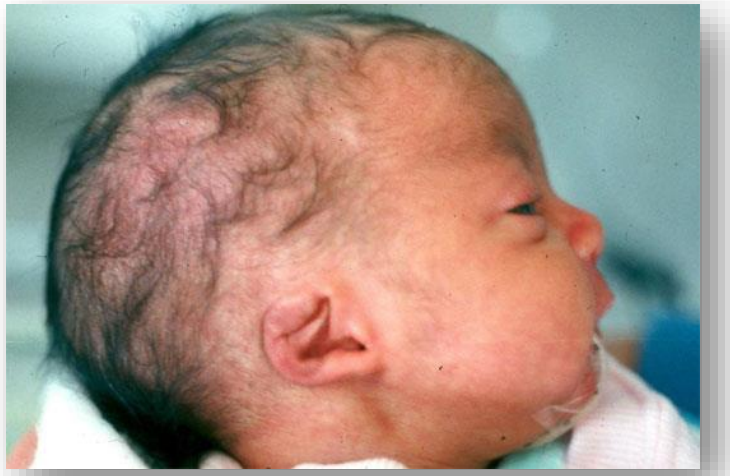
Edwards syndrome

- **What's the name of this sign ?**
  - Rocker bottom feet
- **What's the most likely Diagnosis ?**
  - Edwards syndrome (Trisomy 18)
- **Give other 2 physical findings related to this disorder (not seen in the image)**
  - Microcephaly
  - Ocular hypertelorism
  - Low-set, malformed ears



Edwards syndrome

- **What's the name of this sign ?**
  - Low set and malformed ears
- **What's the most likely Diagnosis ?**
  - Edwards syndrome (Trisomy 18)
- **Give other 2 physical findings related to this disorder (not seen in the image)**
  - Short sternum
  - Ocular hypertelorism
  - Rocker bottom feet



Edwards syndrome

- **What's the name of this sign ?**
  - Clinched fist / Over riding fingers
- **What's the most likely Diagnosis ?**
  - Edwards syndrome (Trisomy 18)
- **Give other 2 another anomies**
  - Short sternum
  - Ocular hypertelorism
  - Rocker bottom feet
  - prominent occiput
- **How to confirm your diagnosis ?**
  - karyotype

The findings in Edwards Syndrome may be remembered with PRINCE Edward:

- **P**rominent occiput
- **R**ocker-bottom feet
- **I**ntellectual disability
- **N**ondisjunction
- **C**lenched fists with overlapping fingers
- low-set **E**ars

شرح

Patau syndrome (trisomy 13)

Epidemiology
Incidence: ~ 1:7400 live births

Etiology
- Free trisomy 13 (75%)
- Trisomy 13 with Robertsonian translocation (20%)

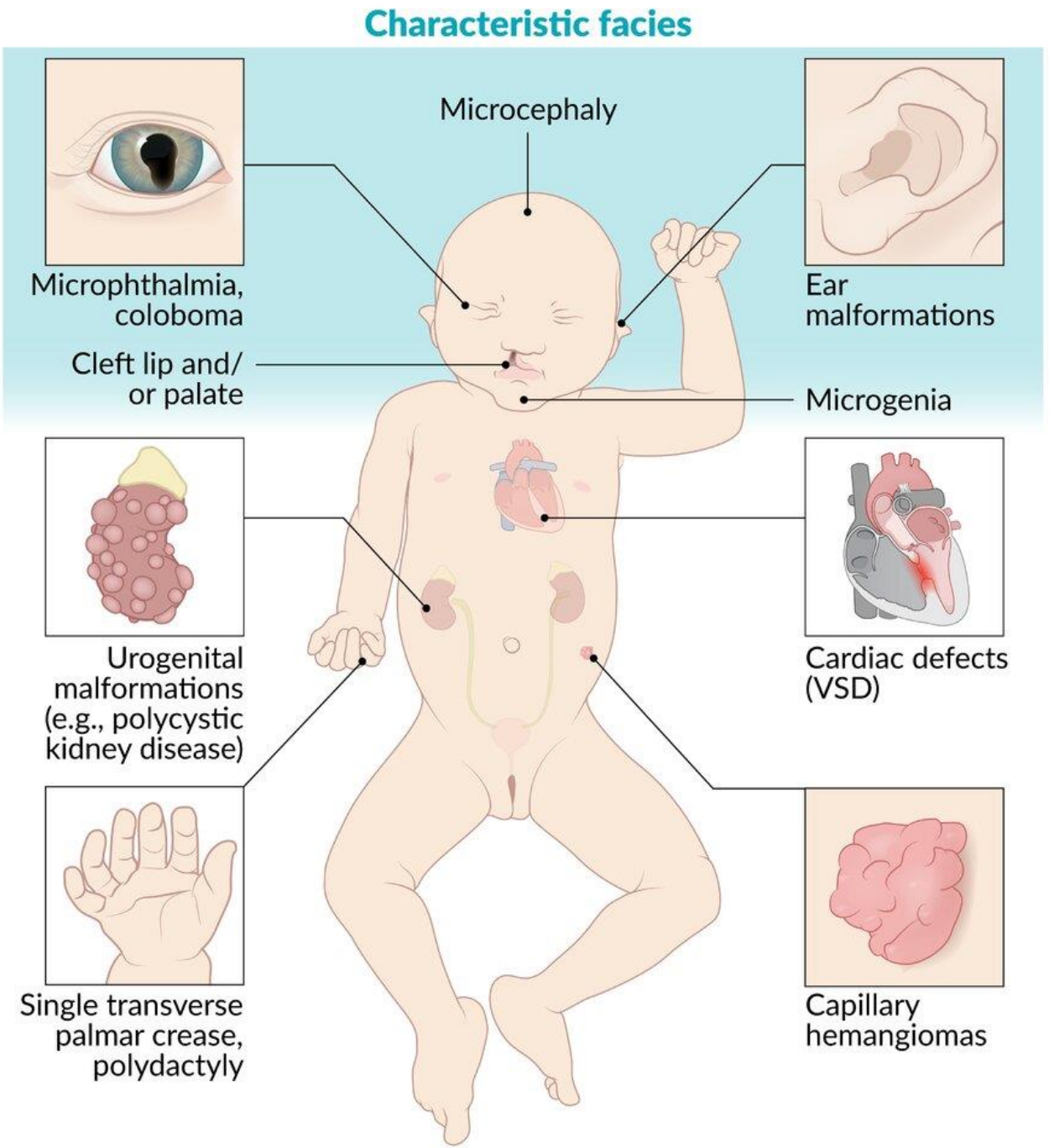
Karyotype
♀ : 47, XX, +13
♂ : 47, XY, +13

Prognosis
Median survival if live-born: ~ 5 days
1 year survival probability: ~ 11%

Note
Correlation with maternal age in free trisomy 13

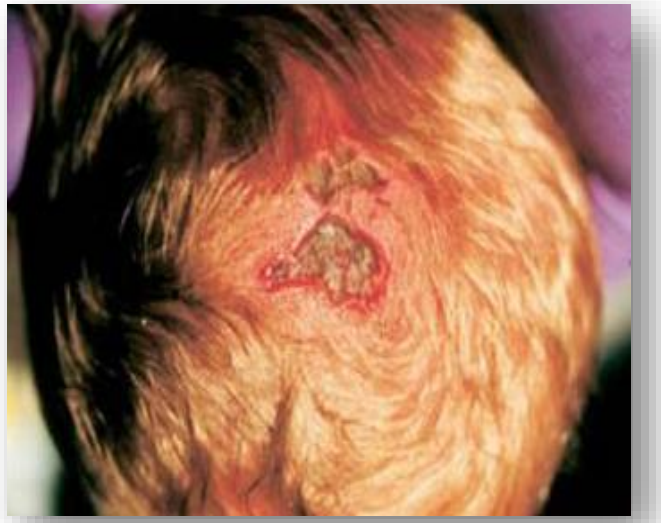
Neurological features

Severe mental and motor impairment



Patau syndrome

- **Name of this lesion**
  - Aplasia cutis congenita
- **What's the name of this syndrome ?**
  - Patau syndrome
- **Mention 2 other findings**
  - Postaxial polydactyly
  - Midline facial defects such as:
    - cyclopia (single orbit)
    - cebocephaly (single nostril)
    - cleft lip and palate



The findings in Patau Syndrome may be remembered with the 5'Ps

- cleft **P**alate
- holo**P**rosencephaly
- **P**olydactyly
- a**P**lasia cutis
- **P**olycystic Kidney disease

شرح

Klinefelter syndrome

Epidemiology
Incidence approx. 1:650 in the US

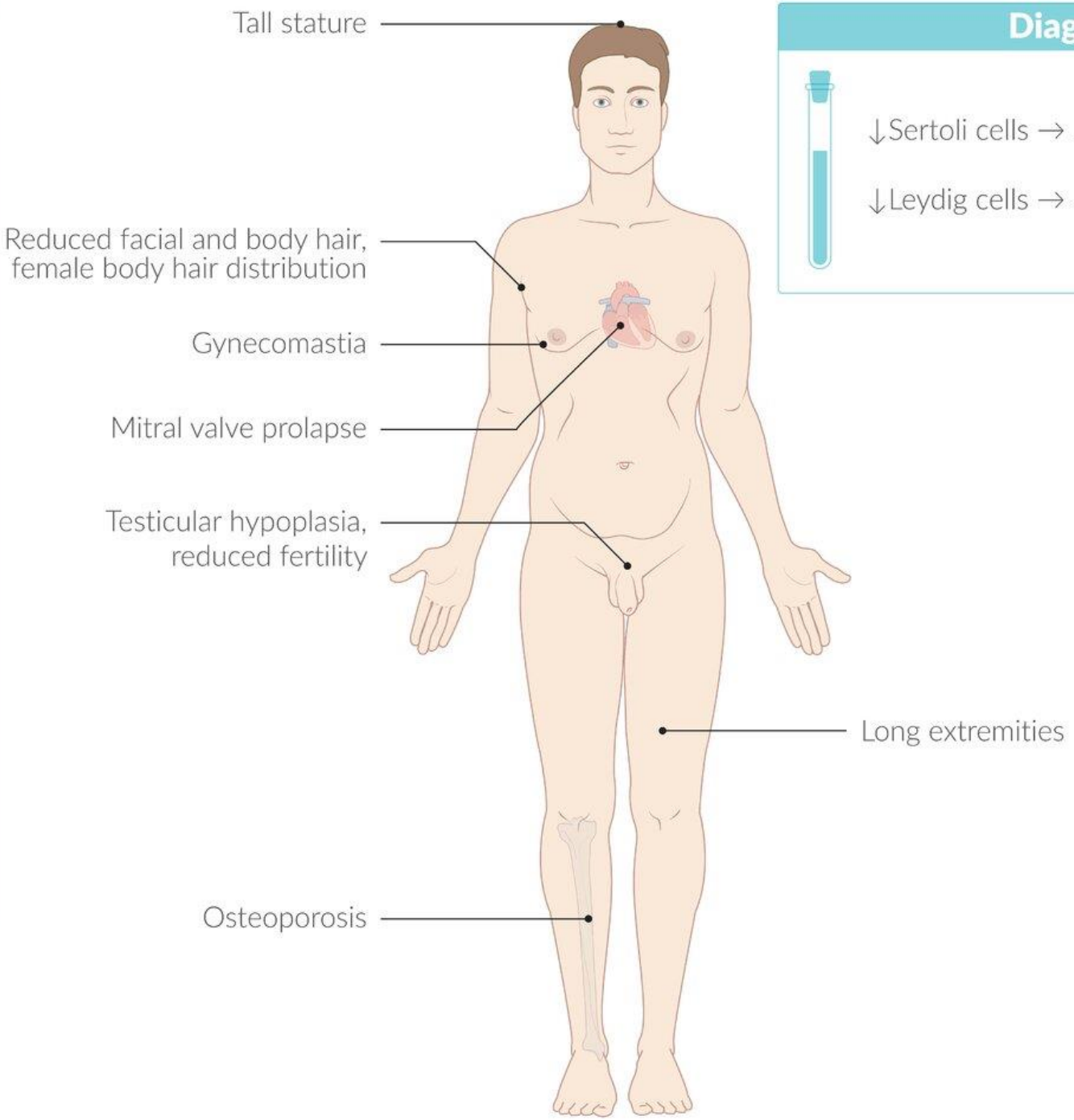
Etiology
Usually due to nondisjunction of sex chromosomes during meiosis.
Associated with an advanced maternal age

Karyotype
47,XXY
Rarely 48,XXXY or 48,XXYY
Barr body is present

Phenotype
Male

Important
Possible developmental delay;
onset of symptoms usually at the start of puberty;
one of the most common causes of male hypogonadism

Karyotype



Diagnosis

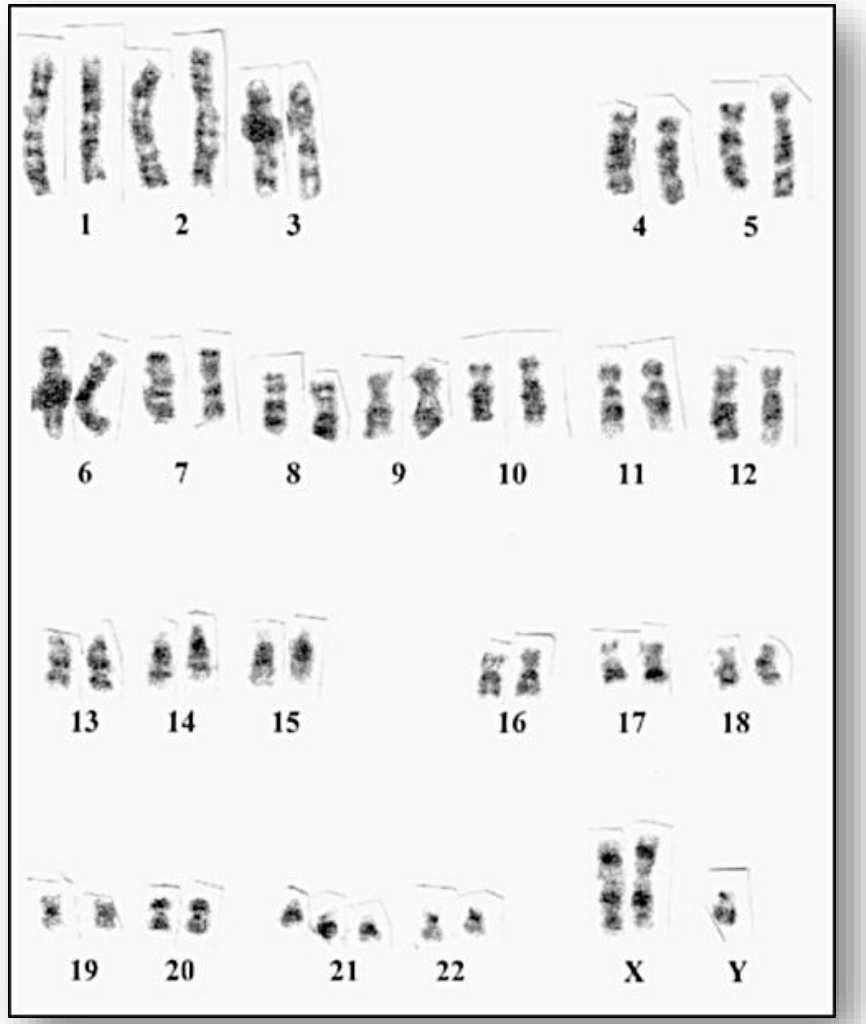
↓Sertoli cells → ↓inhibin B → ↑FSH

↓Leydig cells → ↓testosterone → ↑LH

Klinefelter syndrome

- 15y old male patient presented with delay maturity of the

- **What is your diagnosis ?**
  - Klinefelter syndrome
- **Write 2 physical findings**
  - Gynecomastia + broad hip + tall stature + smaller penis and testicles + less facial and body hair
- **Write 2 abnormalities of this situation**
  - Delayed or incomplete puberty + infertility



شرح

Turner syndrome

Epidemiology
approx. 1/2.500

Etiology
Nondisjunction during meiosis (45,XO) or nondisjunction during mitosis of an embryonic cell

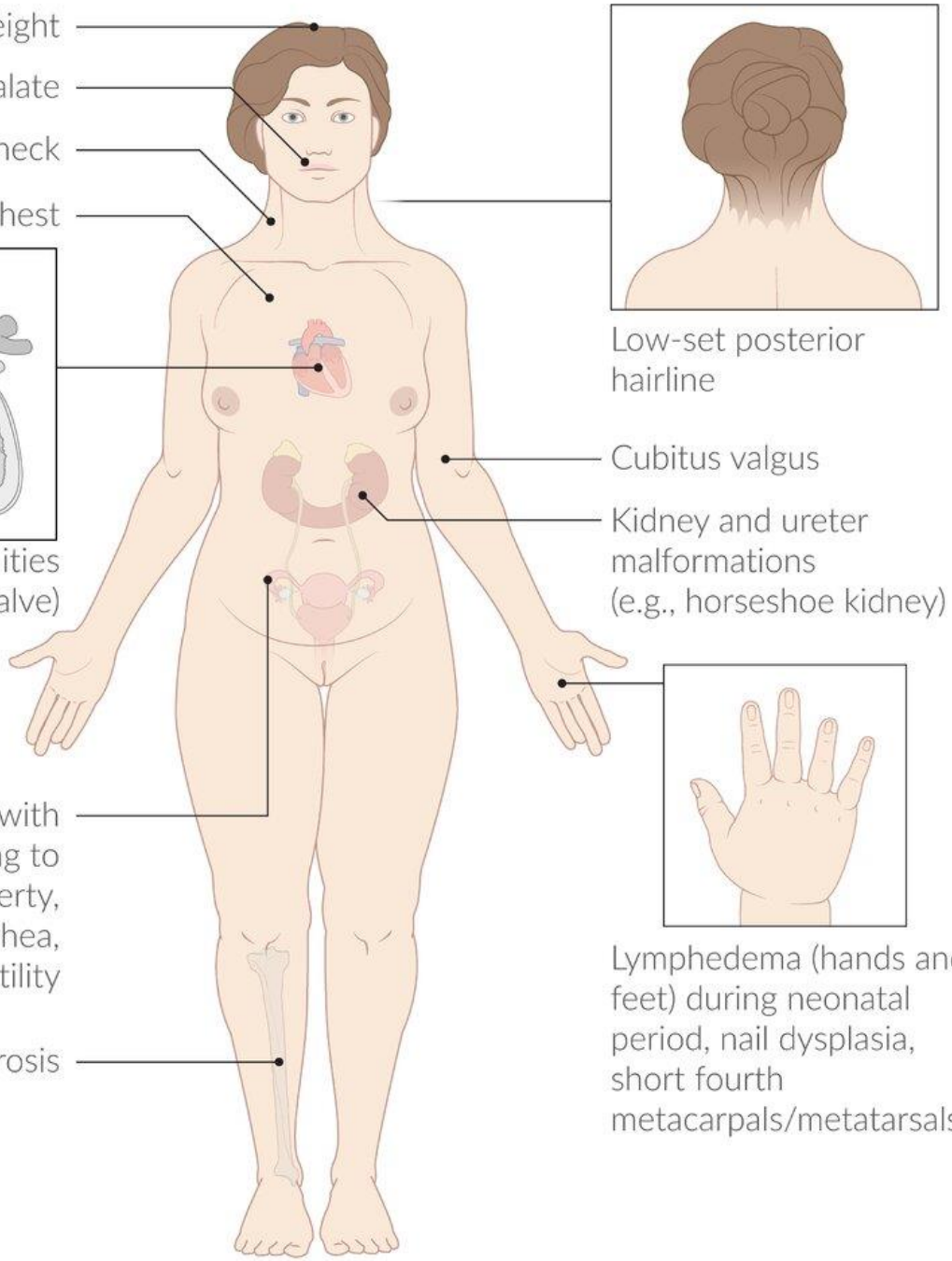
Karyotype
45,XO, female phenotype

Important
Not associated with maternal age

Life expectancy
Reduced by >10 years

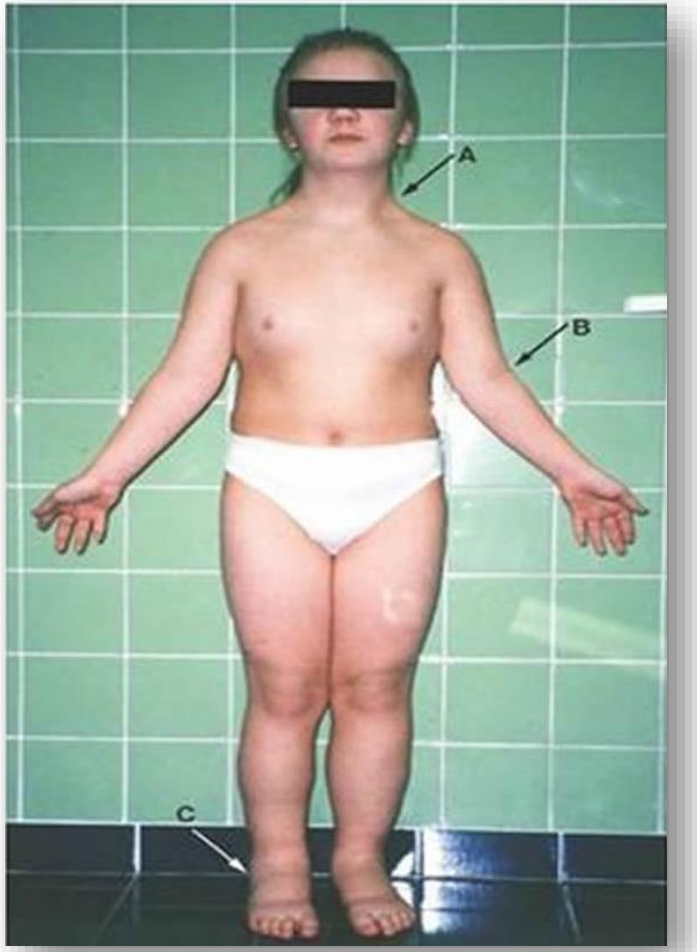
Diagnosis

↓ Estrogen
↓ Androgen
↑ FSH
↑ LH
(Hypergonadotropic hypogonadism)



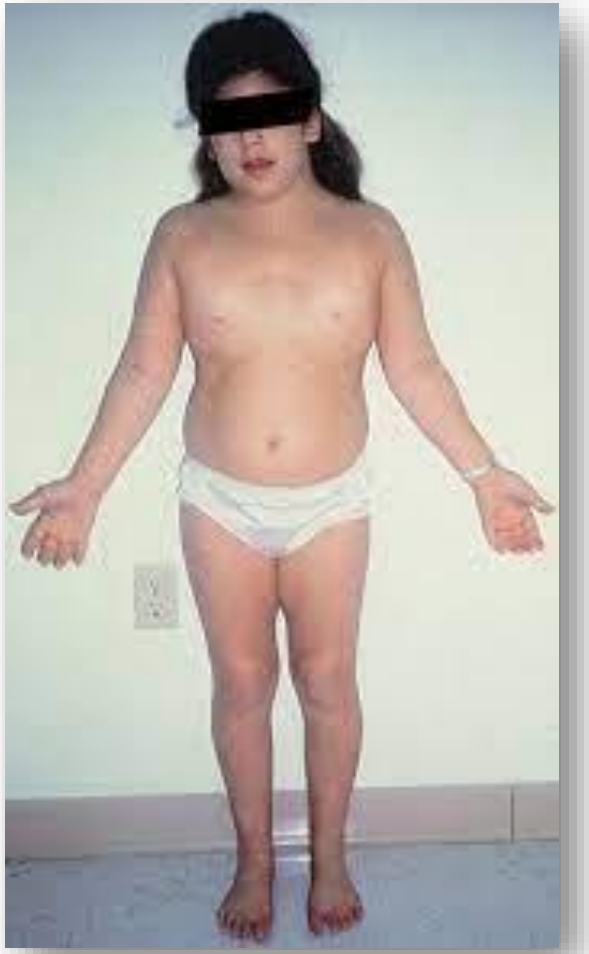
Turner syndrome

- **What is your diagnosis ?**
  - Turner syndrome
- **Mention 2 dysmorphic features**
  - Webbed neck, Short stature
- **What abnormality is seen in the kidneys ?**
  - Horseshoe kidney
- **What future problems will this female have ?**
  - Short stature, infertility
- **What is the karyotyping**
  - 45 XO



Turner syndrome

- **What is your diagnosis ?**
  - Turner syndrome
- **What is the main cardiovascular abnormality in this patient?**
  - Coarctation of the aorta
- **What is the cause of hypertension ?**
  - Coarctation of the aorta
  - Renal anomalies

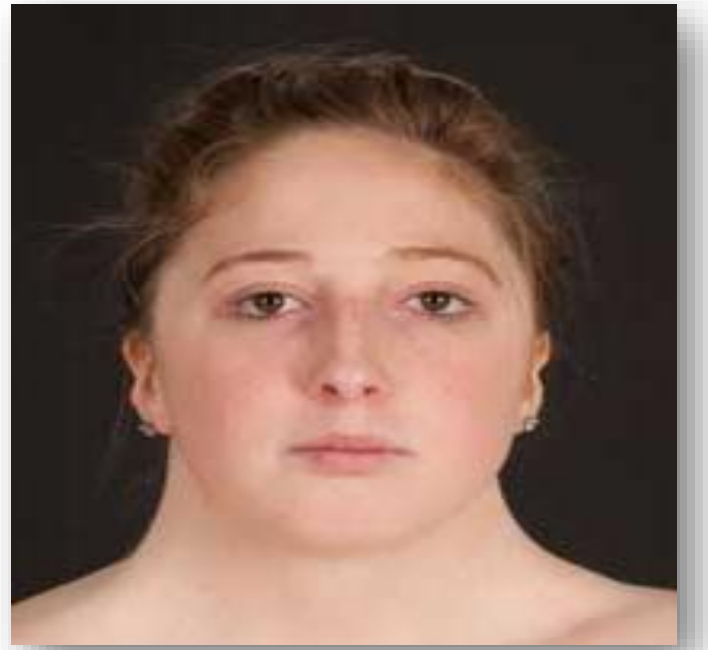


Turner syndrome

- A 16-year-old girl came to your clinic with primary amenorrhea & delayed puberty
- **What is this syndrome, what is the chromosomal pattern?**
  - Turner Syndrome, 45 XO
- **Which cardiac lesion do you want to rule out?**
  - Coarctation of the Aorta

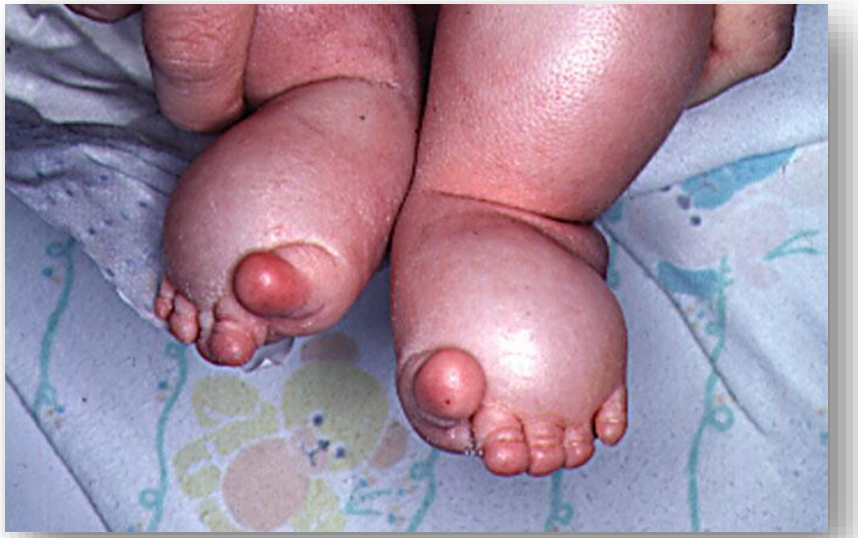
Turner syndrome

- **What is your diagnosis ?**
  - Turner syndrome
- **Mention 2 complications ?**
  - Coarctation of the aorta
  - Renal anomalies



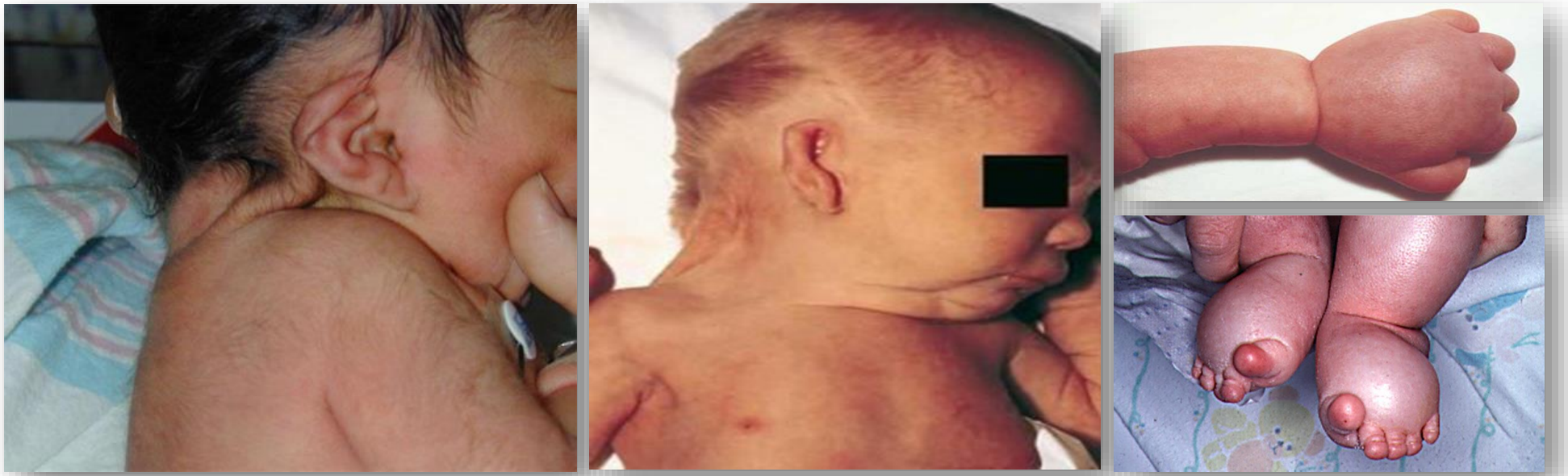
Turner syndrome

- 2-month-old female infant presented to you with non pitting edema
- **Name this sign**
  - Lymphedema
- **What is the syndrome that causes this feature?**
  - Turner syndrome



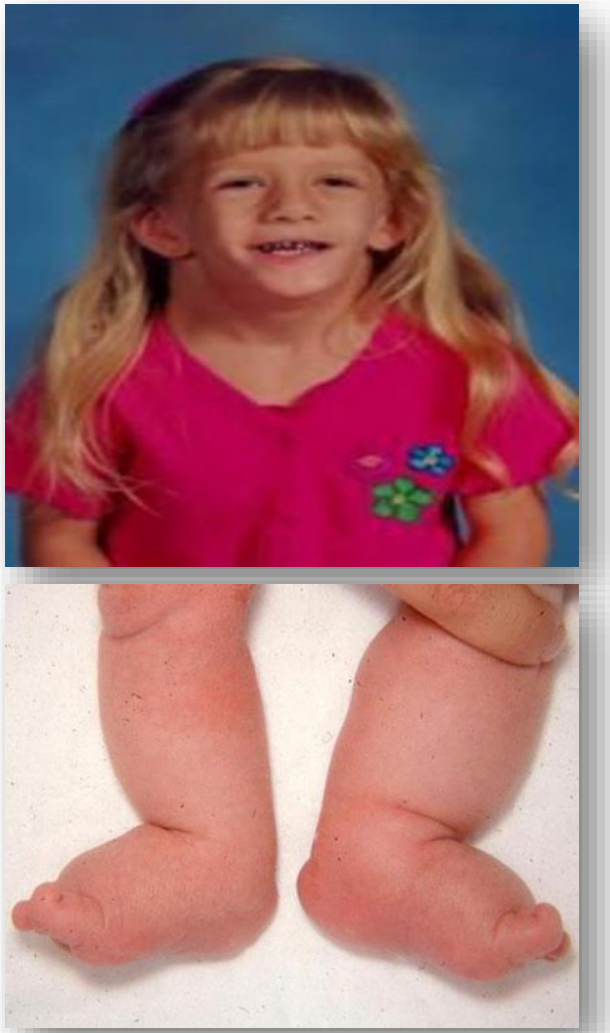
Turner syndrome

- **Mention 3 signs that you can see**
  - Webbed neck, Wide-spaced nipples, Lymphedema of the limbs, Low hair line.
- **Mention Single best test to diagnose**
  - Karyotyping.



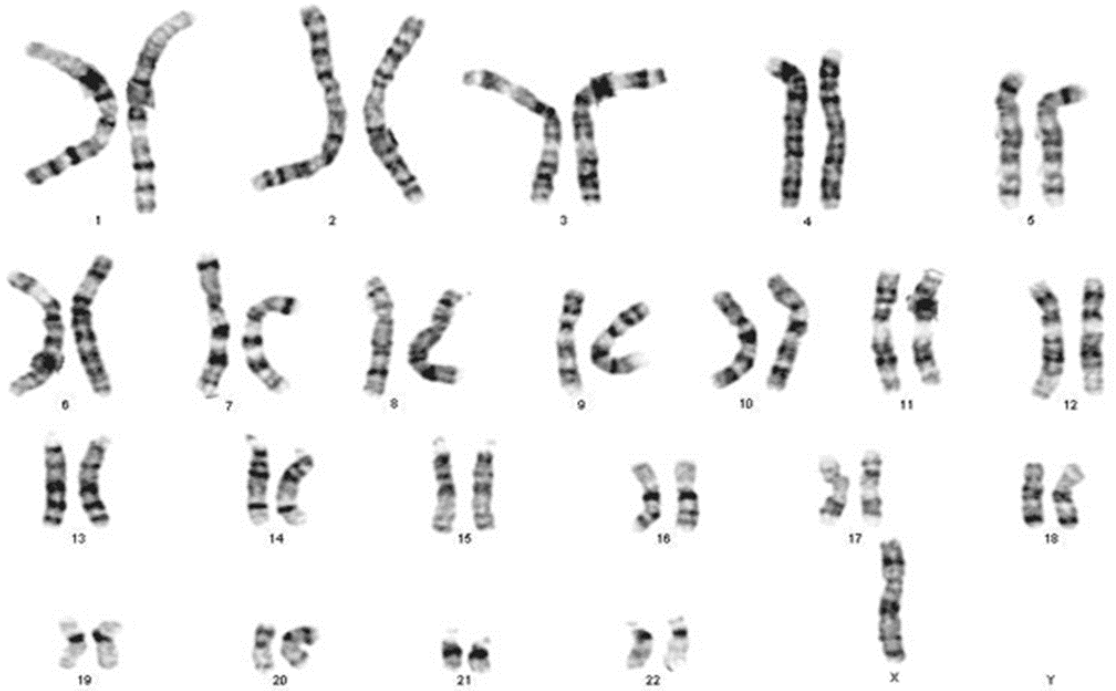
Turner syndrome

- **What's the name of this syndrome ?**
  - Turner syndrome.
- **What's the main CVS abnormality in this girl ?**
  - Coarctation of the aorta (most common).
- **Name other associated congenital defects.**
  - Bicuspid aortic valve; later in life, post-stenotic aortic dilation with aneurysm may develop.
  - Also, renal anomalies e.g., horseshoe kidney.



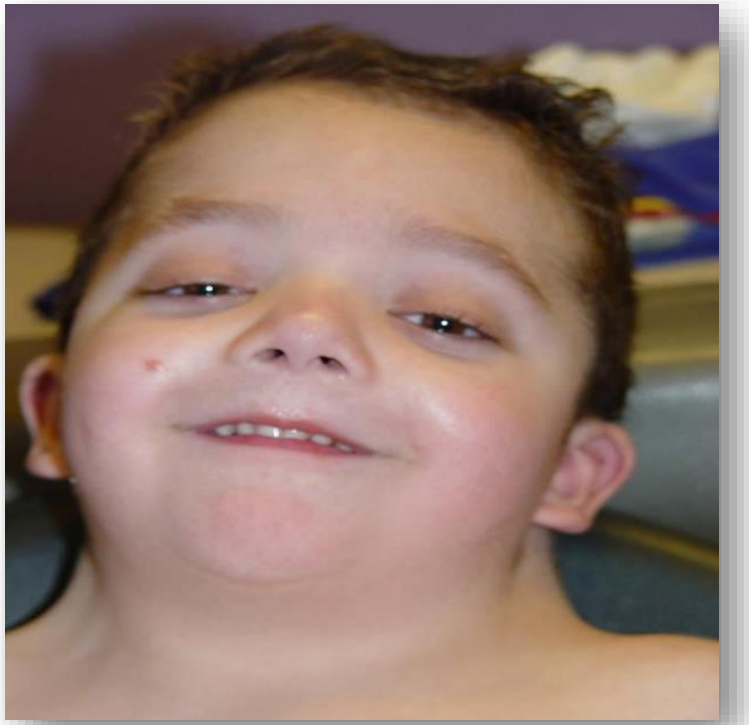
This Karyotype indicates which syndrome

- Turner Syndrome, 45 XO



Noonan’s Syndrome

- **What’s the name of this syndrome ?**
  - Noonan’s Syndrome .
- **What’s the chromosomal defect here ?**
  - Autosomal dominant (AD) congenital disorder.
- **What’s the male:female ratio of incidence ?**
  - 1 : 1 … as it’s AD.



➢Sometimes; described as “the male version of Turner's syndrome”

شرح

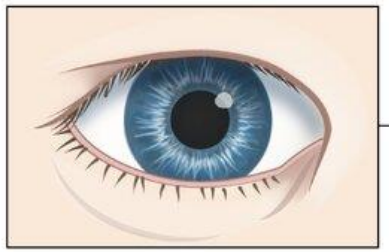
Williams syndrome

Epidemiology
1/10,000 live births

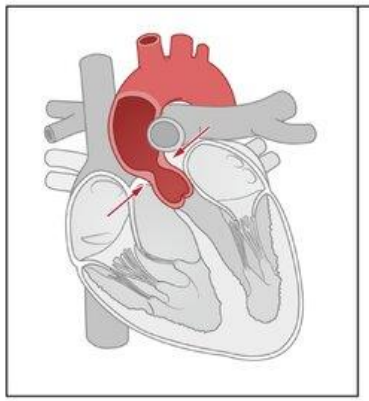
Etiology
Microdeletion on chromosome 7
(includes deletion of elastin gene)
Mostly spontaneous mutation, rarely
autosomal-dominant Inheritance

Laboratory findings
Intermittent hypercalcemia,
mainly during early childhood
(due to increased vitamin D sensitivity)

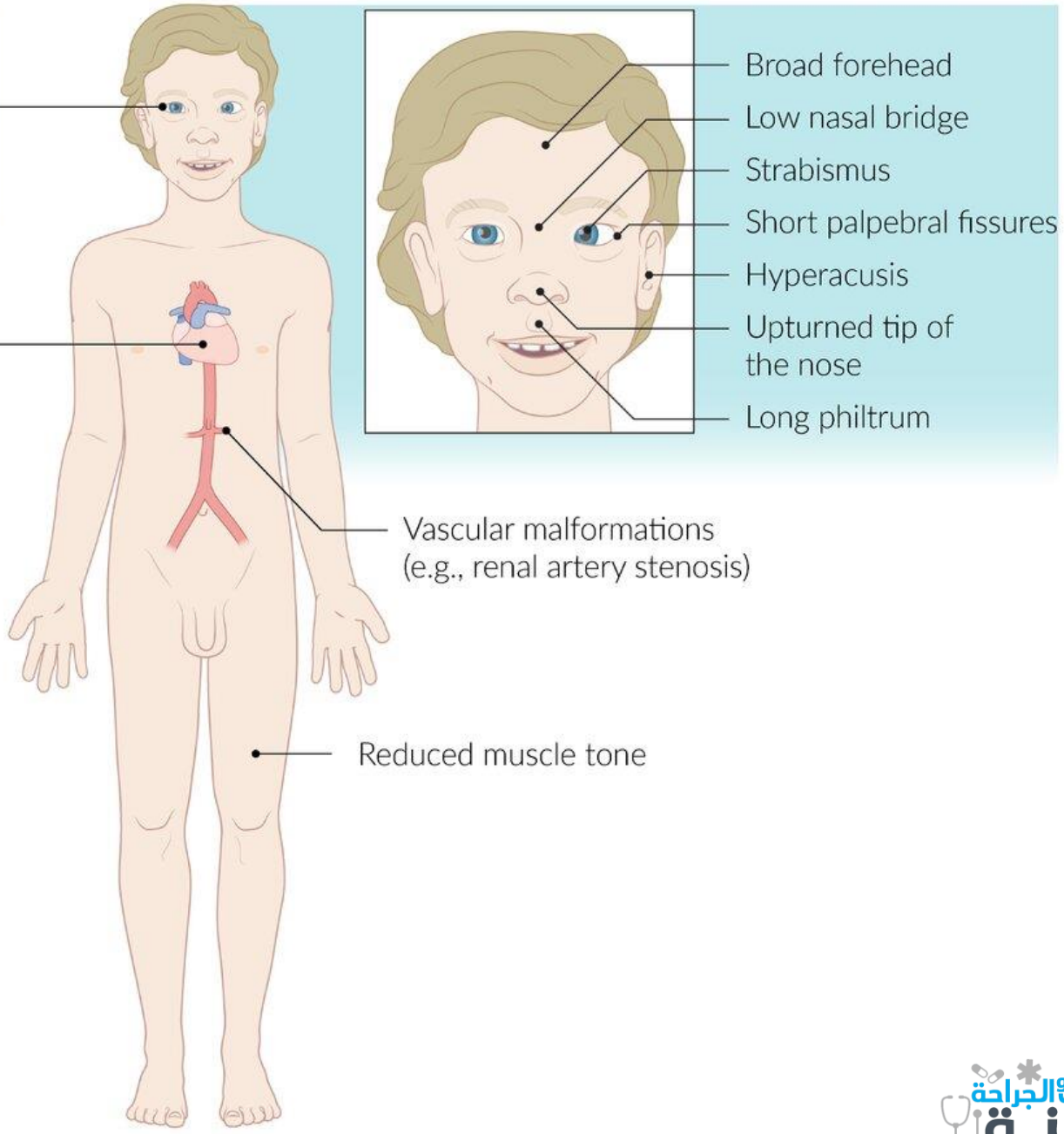
Complications
Mostly cardiovascular
Gastrointestinal symptoms, vomiting
and, subsequently,
failure to thrive



Stellate pattern in the iris



Cardiac malformations
(esp. supravalvular
aortic stenosis)

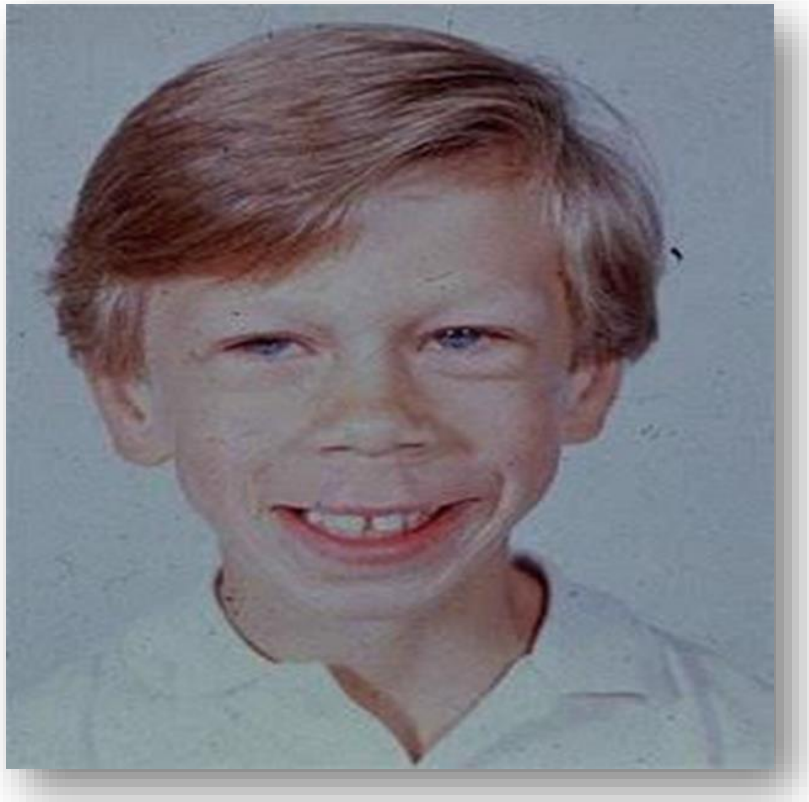


Psychiatric manifestations

Cognitive deficits
Typically sociable personality with good verbal skills
(cocktail party personality)
Anxiety disorders, phobias

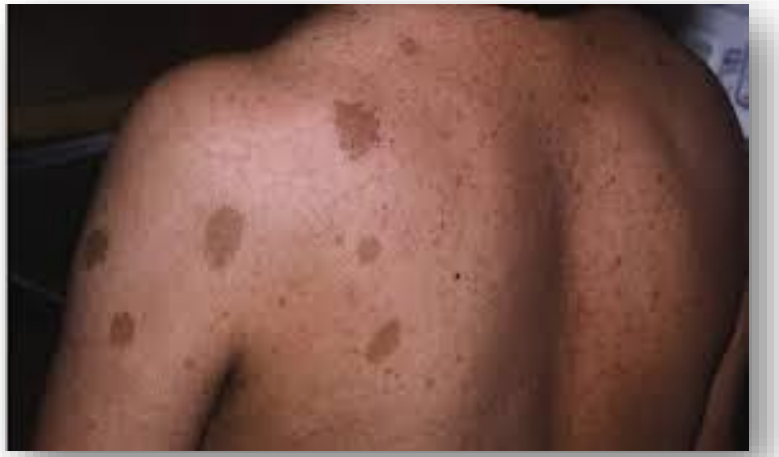
Williams syndrome

- **What's the name of this syndrome ?**
  - William 's syndrome
- **What's the chromosomal defect here ?**
  - A small deletion of chromosome 7q11.
- **Name 2 associated congenital defects**
  - Supra-valvular aortic stenosis & pulmonic stenosis & peripheral pulmonic stenosis.



McCune–Albright syndrome

- **What is the name of this finding ?**
  - Unilateral café-au-lait spots with unilateral, ragged edges
- **if it is associated with precocious, what is the dx ?**
  - McCune–Albright syndrome



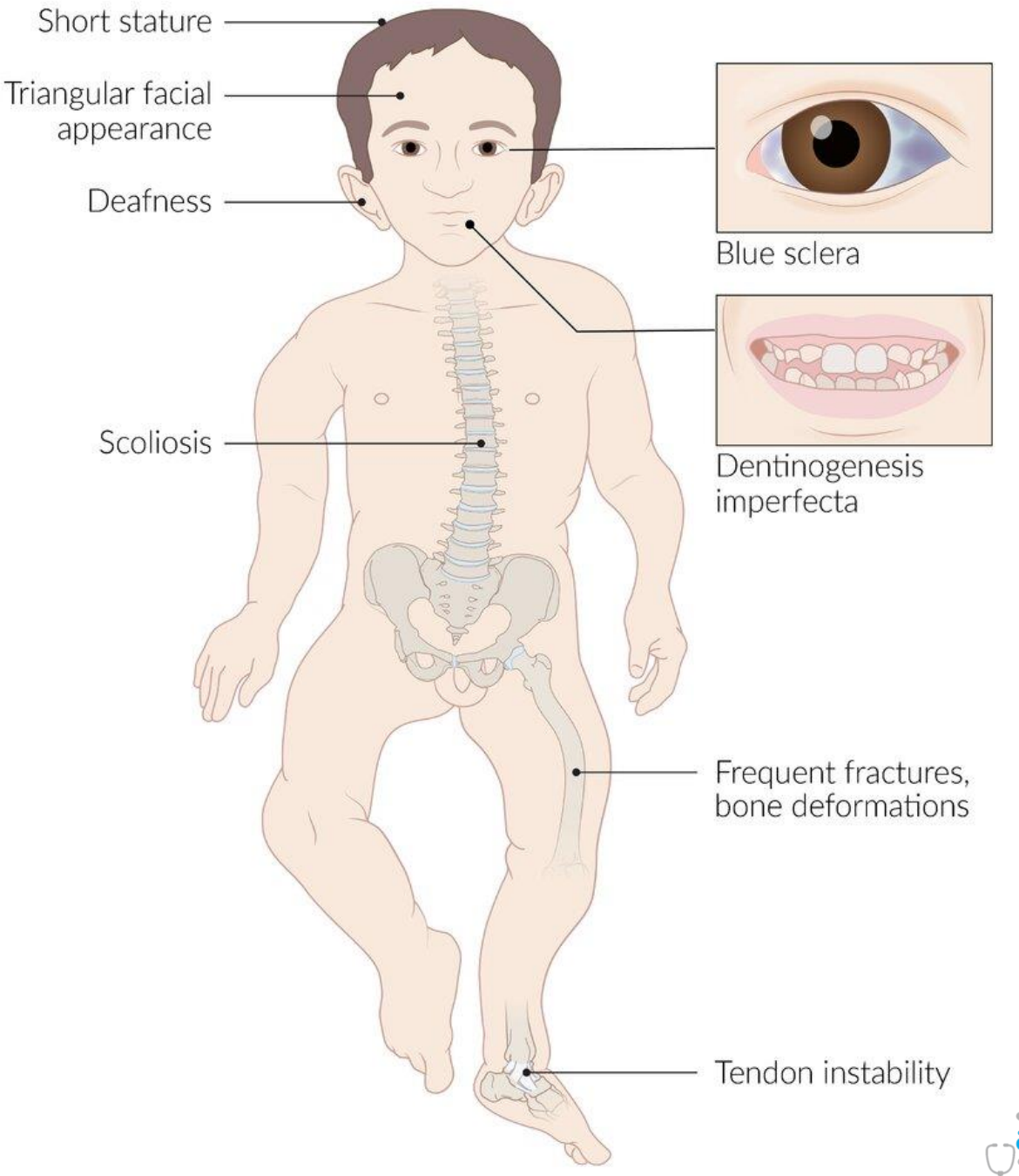
Osteogenesis imperfecta

Epidemiology
Prevalence: Approx. 1/10,000–20,000
♀ = ♂

Classification
Eleven types with highly variable genotypes and phenotypes

Etiology
Disorder of collagen type I synthesis; inheritance can be autosomal dominant (most common) but also autosomal recessive (rare)

Life expectancy
Mild forms: typically normal
Very severe form (type II): typically lethal within the first year of life

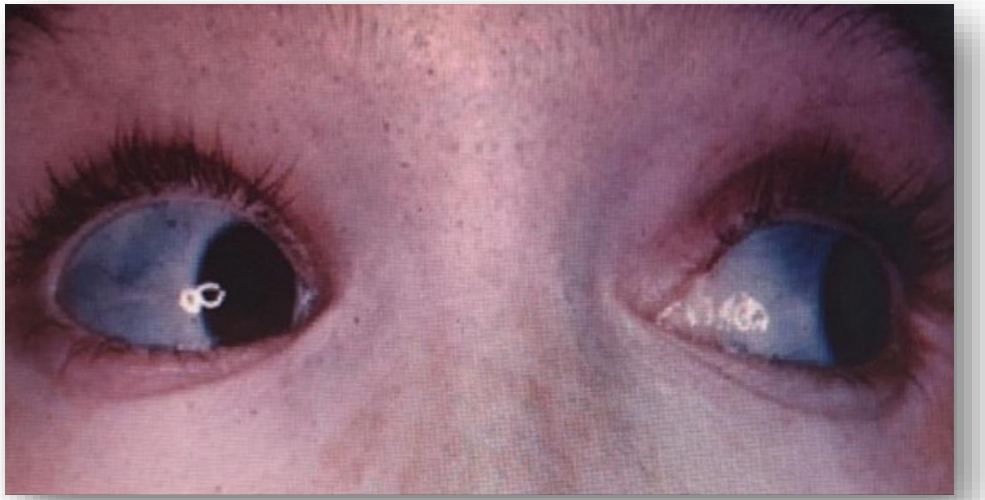


Osteogenesis imperfecta

➢ The eyes of 2-year-old boy baby presented with short stature.

❖ **What other clues in the history will help you reach the diagnosis ?**

a. Proportionate short stature
b. Multiple bone fracture
c. Family history of the same condition
d. Presence of wide arm span
e. Breech presentation



Prader-Willi syndrome

Epidemiology
Prevalence ~ 1 in 16000 - 25000
♂ = ♀

Etiology
Paternal deletion 15q11q13 (> 70%)
Maternal uniparental disomy
Low risk of inheritance (mostly spontaneous mutation)
Etiology similar to that of Angelman syndrome

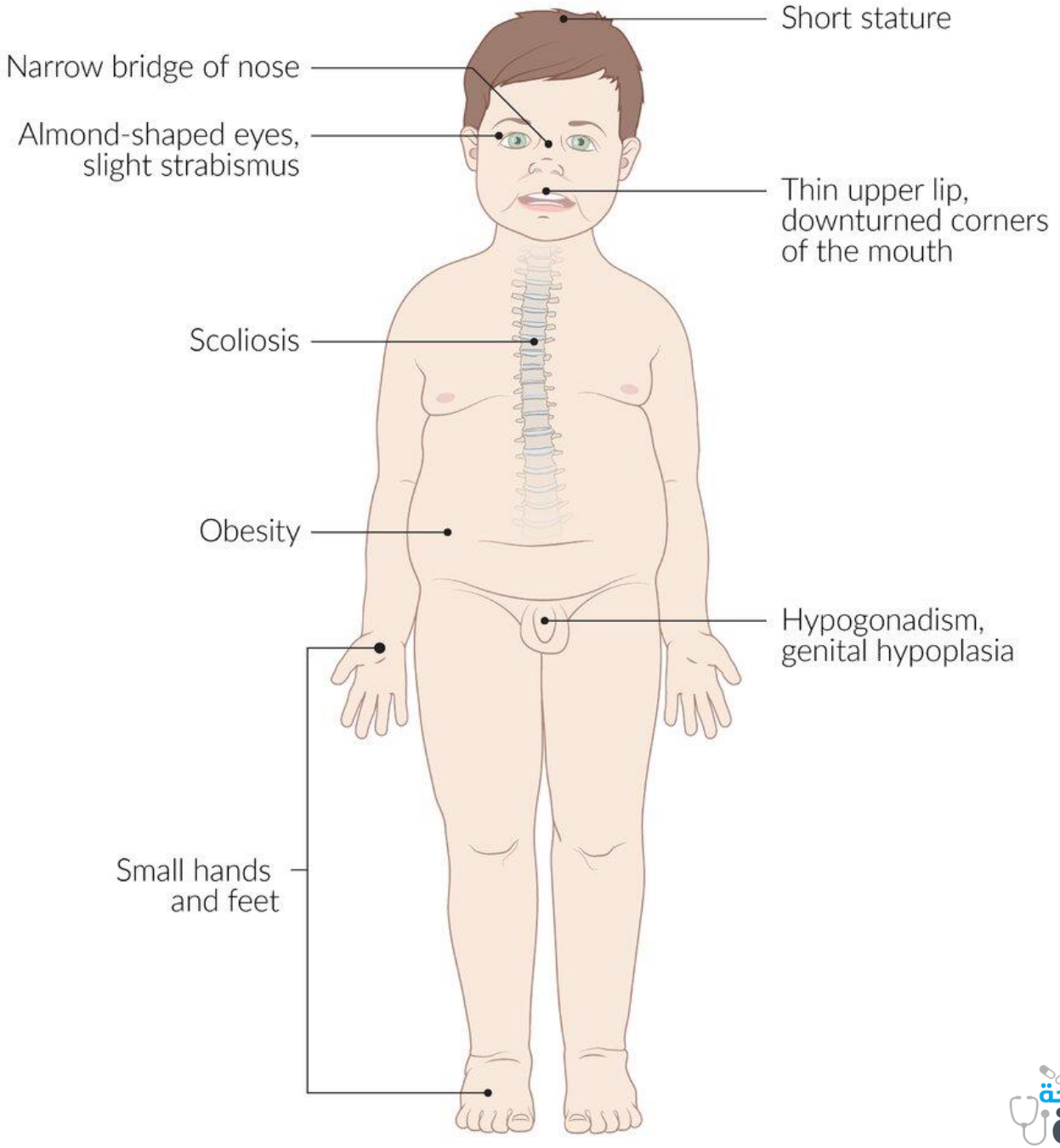
Complications
Obesity and sequelae, e.g., diabetes mellitus and respiratory disorders

Prognosis
Partial autonomy is possible

Life expectancy
Slightly below normal if obesity can be controlled

Psychiatric and neurological features

Uncontrolled appetite with hyperphagia, learning difficulties, cognitive deficits, low impulse control, defiant behavior, psychosis



Fragile X syndrome

Epidemiology
Approx. 1 in 8000 females and 1 in 4000 males
Second most common cause of congenital cognitive disorders after trisomy 21

Etiology
Mutation of the FMR1 gene
(CGG trinucleotide repeat expansion)
X-linked inheritance

Complications
Epilepsy

Note
Milder manifestations in heterozygous female
(mostly neuropsychiatric disorders, rarely cognitive deficits)

Life expectancy
Typically normal

Characteristic facies

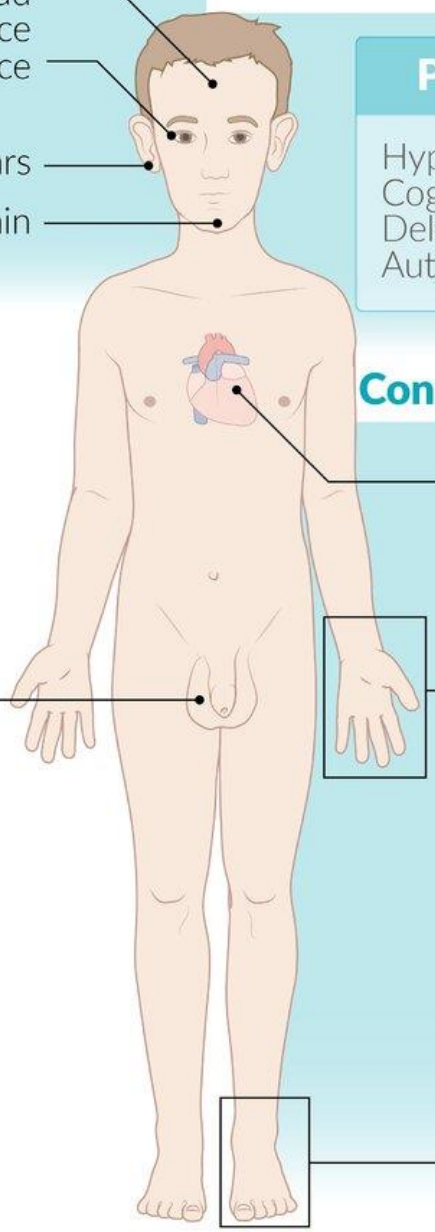
Prominent forehead and increased head circumference
Long, slim face
Large ears
Prominent, pointy chin

Psychiatric features

Hyperactivity
Cognitive deficits
Delayed speech development
Autistic behavior

Connective tissue weakness

Mitral valve prolapse
Testicular enlargement
Hypermobile joints
Pes planus



Cri-du-chat syndrome

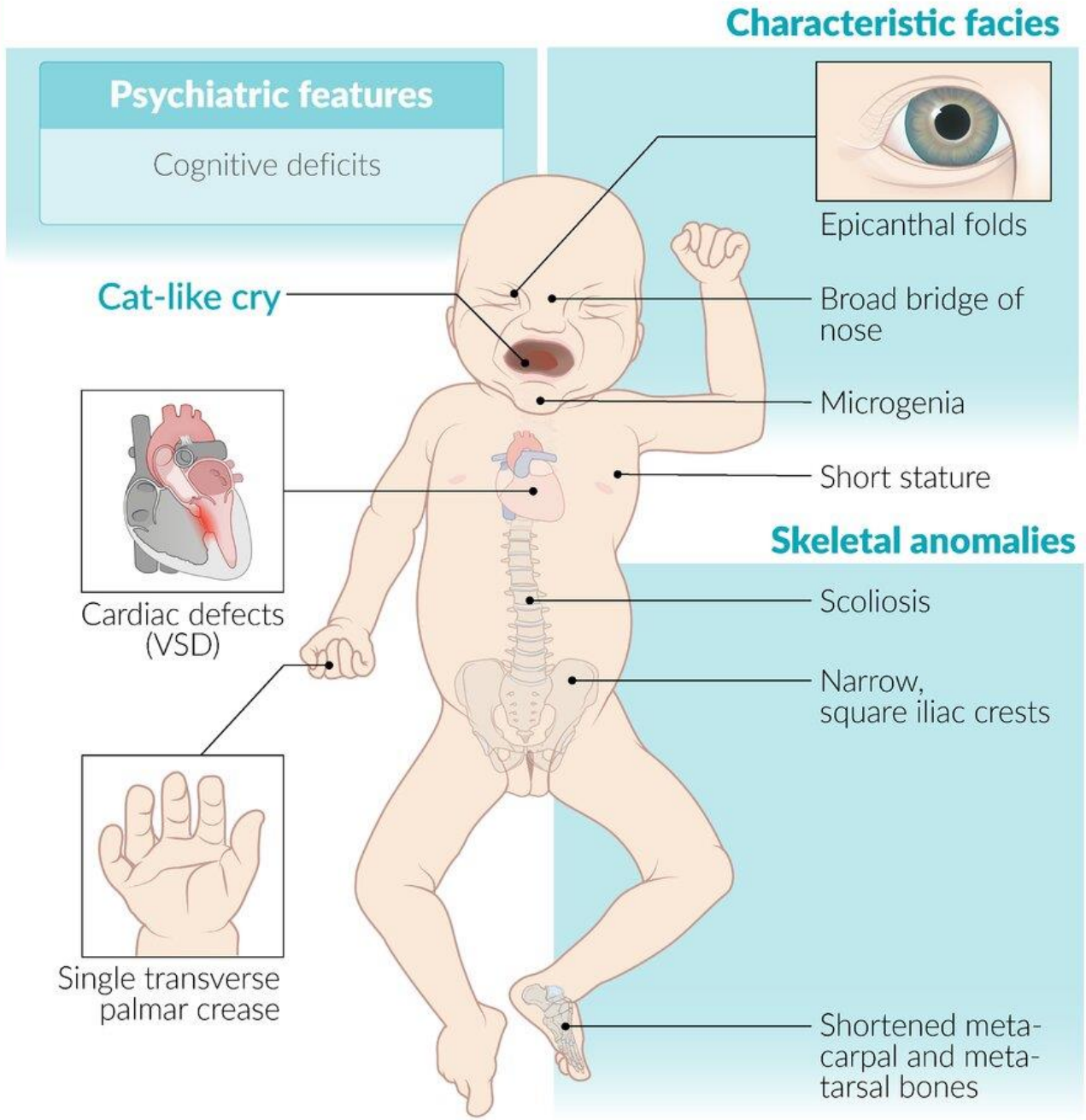
Epidemiology
Approximately 1:45,000 liveborn infants
Sex: ♀ > ♂ (2:1)

Etiology
Structural aberration of the short arm of chromosome 5

Karyotype
♀: 46,XX -5p
♂: 46,XY -5p

Note
Cardinal symptom is a cat-like cry during infancy

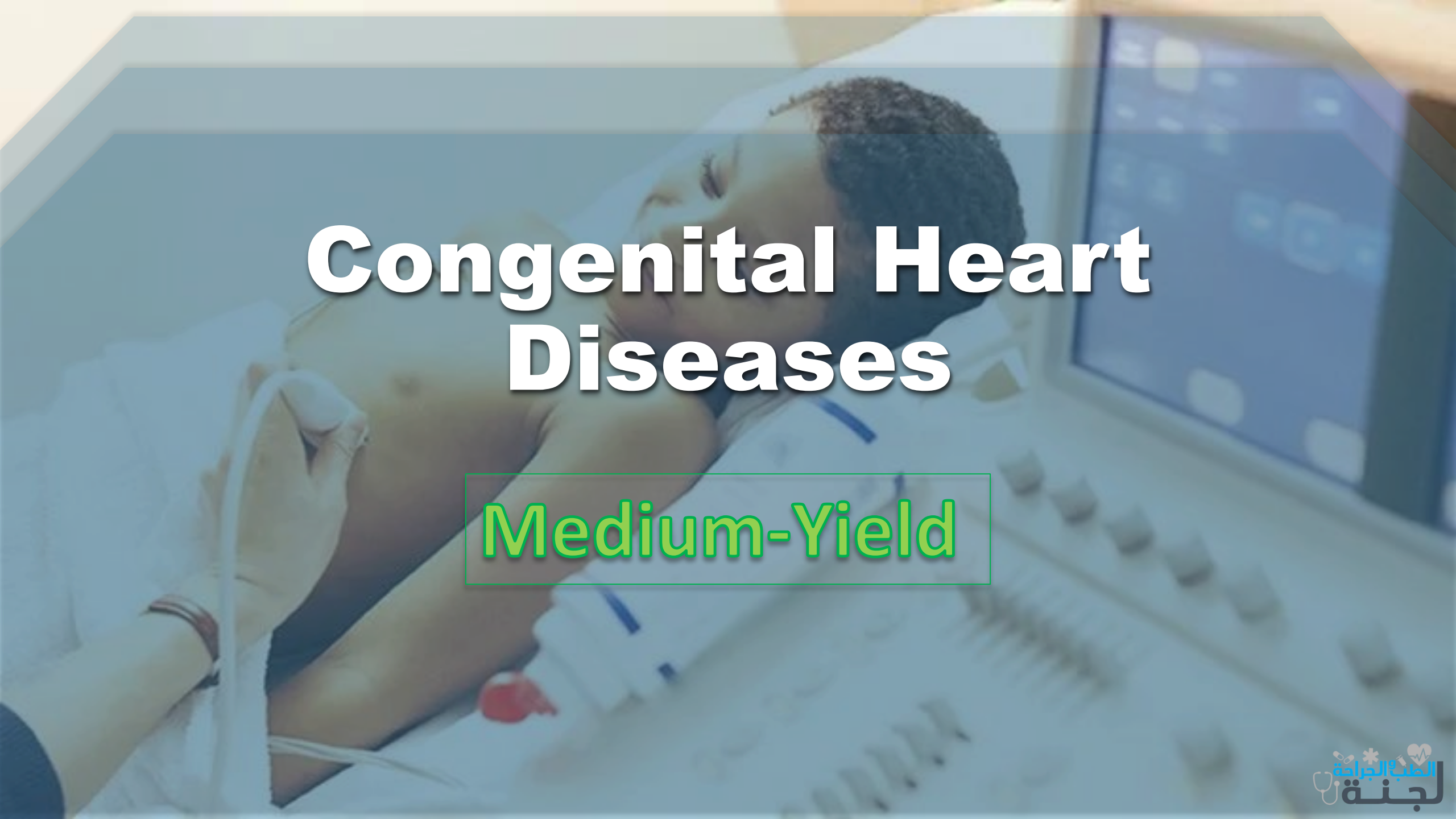
Life expectancy
May be normal, depending on severity of symptoms and treatment



إضافي

Congenital cardiac defect associations

ASSOCIATION	DEFECT
Prenatal alcohol exposure (fetal alcohol syndrome)	VSD, PDA, ASD, tetralogy of Fallot
Congenital rubella	PDA, pulmonary artery stenosis, septal defects
Down syndrome	AV septal defect (endocardial cushion defect), VSD, ASD
Infant of patient with diabetes during pregnancy	Transposition of great vessels, truncus arteriosus, tricuspid atresia, VSD
Marfan syndrome	MVP, thoracic aortic aneurysm and dissection, aortic regurgitation
Prenatal lithium exposure	Ebstein anomaly
Turner syndrome	Bicuspid aortic valve, coarctation of aorta
Williams syndrome	Supravalvular aortic stenosis
22q11 syndromes	Truncus arteriosus, tetralogy of Fallot



Congenital Heart Diseases

Medium-Yield

Congenital Heart Diseases

إضافي

Cyanotic

RIGHT-TO-LEFT SHUNTS

Early cyanosis—"blue babies." Often diagnosed prenatally or become evident immediately after birth. Usually require urgent surgical treatment and/or maintenance of a PDA.

The 5 T's:

1. Truncus arteriosus (1 vessel)
2. Transposition (2 switched vessels)
3. Tricuspid atresia (3 = Tri)
4. Tetralogy of Fallot (4 = Tetra)
5. TAPVR (5 letters in the name)

Acyanotic

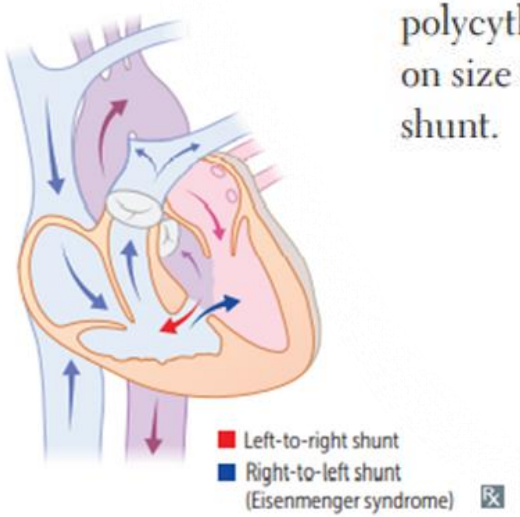
LEFT-TO-RIGHT SHUNTS

Acyanotic at presentation; cyanosis may occur years later. Frequency: VSD > ASD > PDA.

Right-to-left shunts: early cyanosis.
Left-to-right shunts: "later" cyanosis.

Eisenmenger syndrome

Uncorrected left-to-right shunt (VSD, ASD, PDA) → ↑ pulmonary blood flow → pathologic remodeling of vasculature → pulmonary arterial hypertension. RVH occurs to compensate → shunt becomes right to left when RV > LV pressure (see illustration). Causes late cyanosis, clubbing, and polycythemia. Age of onset varies depending on size and severity of initial left-to-right shunt.



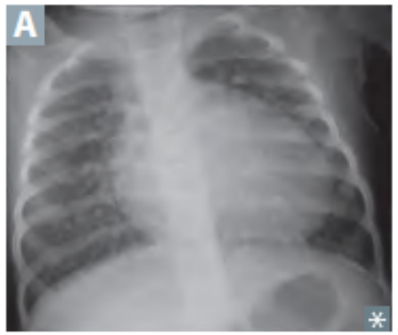
إضافي

Congenital Heart Diseases – Cyanotic

Persistent truncus arteriosus

Truncus arteriosus fails to divide into pulmonary trunk and aorta due to failure of aorticopulmonary septum formation; most patients have accompanying VSD.

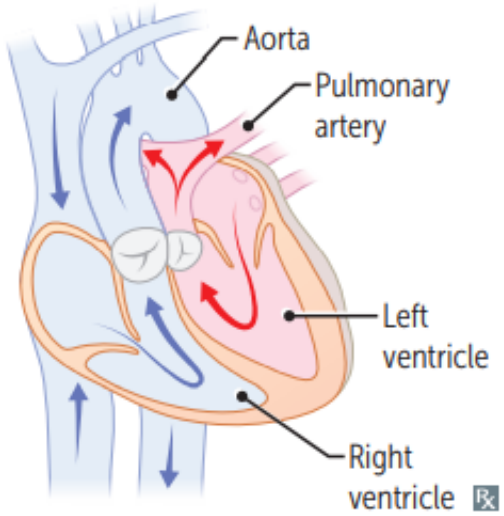
D-transposition of great arteries



Aorta leaves RV (anterior) and pulmonary trunk leaves LV (posterior) → separation of systemic and pulmonary circulations. Not compatible with life unless a shunt is present to allow mixing of blood (eg, VSD, PDA, or patent foramen ovale).

Due to failure of the aorticopulmonary septum to spiral ("egg on a string" appearance on CXR) A.

Without surgical intervention, most infants die within the first few months of life.



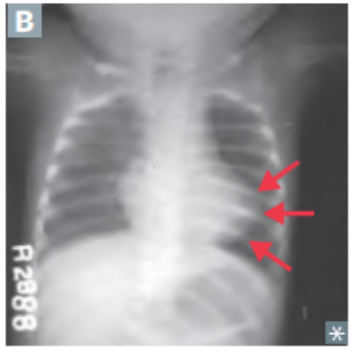
Tricuspid atresia

Absence of tricuspid valve and hypoplastic RV; requires both ASD and VSD for viability.

إضافي

Congenital Heart Diseases – Cyanotic

Tetralogy of Fallot



Caused by anterosuperior displacement of the infundibular septum. Most common cause of early childhood cyanosis.

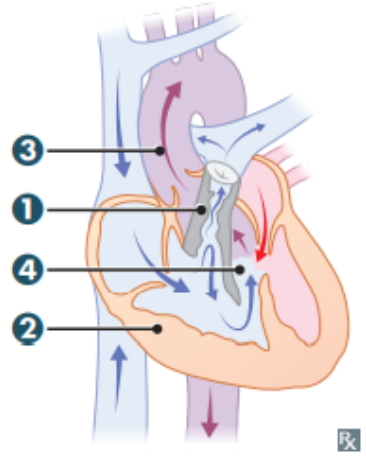
1. **P**ulmonary infundibular stenosis (most important determinant for prognosis)
2. **R**ight ventricular hypertrophy (RVH)—boot-shaped heart on CXR B
3. **O**verriding aorta
4. **V**SD

Pulmonary stenosis forces right-to-left flow across VSD → RVH, "tet spells" (often caused by crying, fever, and exercise due to exacerbation of RV outflow obstruction).

PROVe.

Squatting: ↑ SVR, ↓ right-to-left shunt, improves cyanosis.

Associated with 22q11 syndromes.



Total anomalous pulmonary venous return

Pulmonary veins drain into right heart circulation (SVC, coronary sinus, etc); associated with ASD and sometimes PDA to allow for right-to-left shunting to maintain CO.

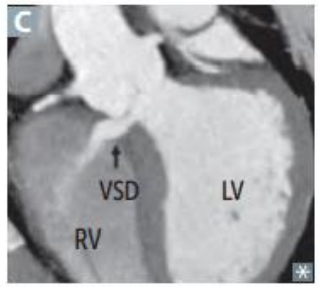
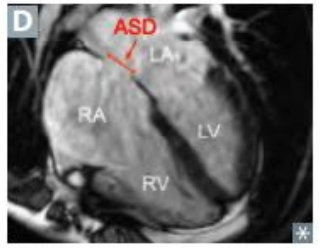
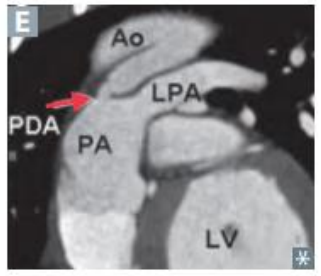
Ebstein anomaly

Displacement of tricuspid valve leaflets downward into RV, artificially "atrializing" the ventricle. Associated with tricuspid regurgitation, accessory conduction pathways, right-sided HF.

Can be caused by lithium exposure in utero.

إضافي

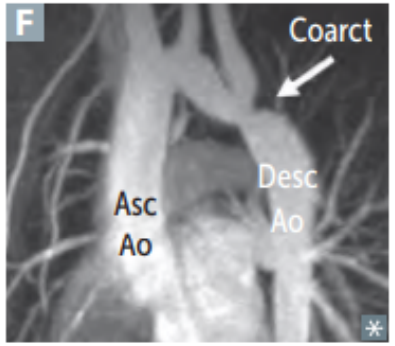
Congenital Heart Diseases – Acyanotic

Ventricular septal defect 	Asymptomatic at birth, may manifest weeks later or remain asymptomatic throughout life. Most self resolve; larger lesions C may lead to LV overload and HF.	O_2 saturation ↑ in RV and pulmonary artery.
Atrial septal defect 	Defect in interatrial septum D; wide, fixed split S2. Ostium secundum defects most common and usually an isolated finding; ostium primum defects rarer and usually occur with other cardiac anomalies. Symptoms range from none to HF. Distinct from patent foramen ovale, which is due to failed fusion.	O_2 saturation ↑ in RA, RV, and pulmonary artery. May lead to paradoxical emboli (systemic venous emboli use ASD to bypass lungs and become systemic arterial emboli). Associated with Down syndrome.
Patent ductus arteriosus 	In fetal period, shunt is right to left (normal). In neonatal period, ↓ pulmonary vascular resistance → shunt becomes left to right → progressive RVH and/or LVH and HF. Associated with a continuous, "machinelike" murmur. Patency is maintained by PGE synthesis and low O_2 tension. Uncorrected PDA E can eventually result in late cyanosis in the lower extremities (differential cyanosis).	PDA is normal in utero and normally closes only after birth.

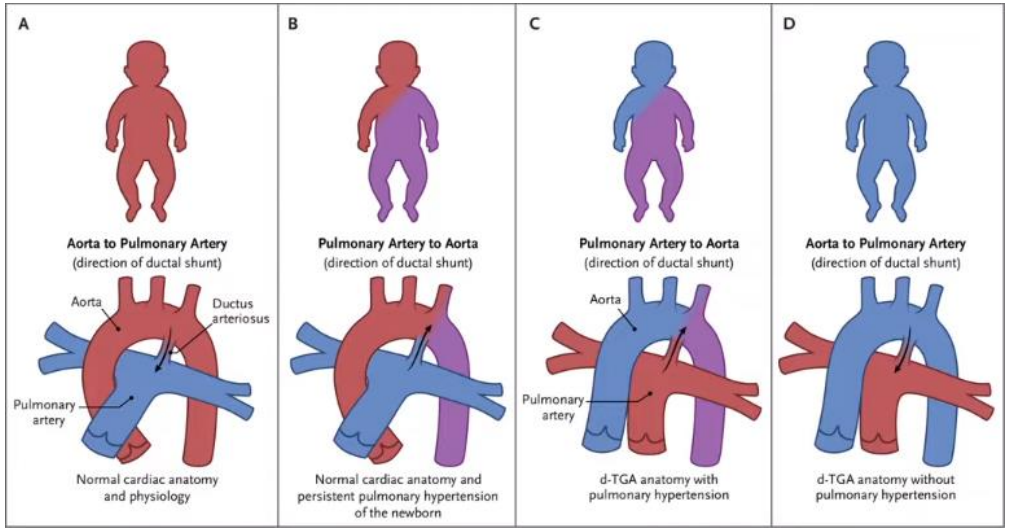
إضافي

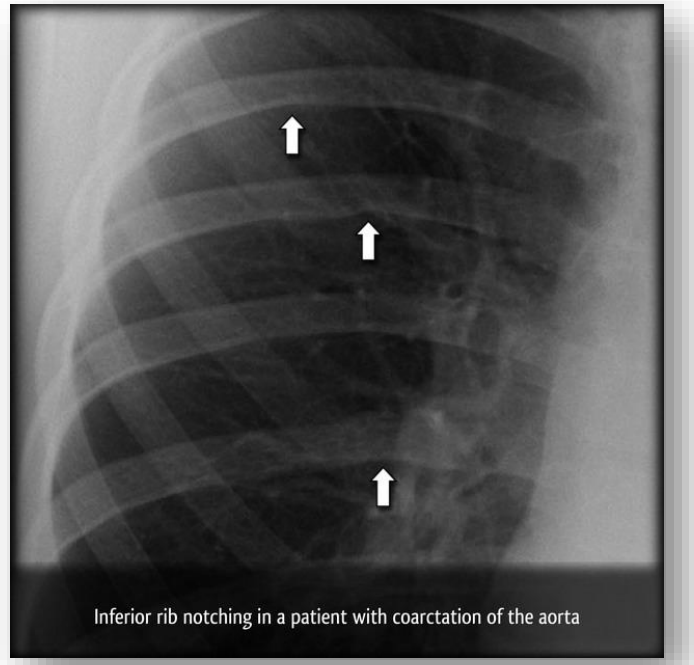
Congenital Heart Diseases – Other anomalies

Coarctation of the aorta



Aortic narrowing F near insertion of ductus arteriosus ("juxtaductal"). Associated with bicuspid aortic valve, other heart defects, and Turner syndrome. Hypertension in upper extremities. Lower extremities are cold with weak, delayed pulses (brachiofemoral delay). With age, intercostal arteries enlarge due to collateral circulation; arteries erode ribs → notched appearance on CXR. Complications include HF, ↑ risk of cerebral hemorrhage (berry aneurysms), aortic rupture, and possible infective endocarditis.




Inferior rib notching in a patient with coarctation of the aorta

Early cyanosis

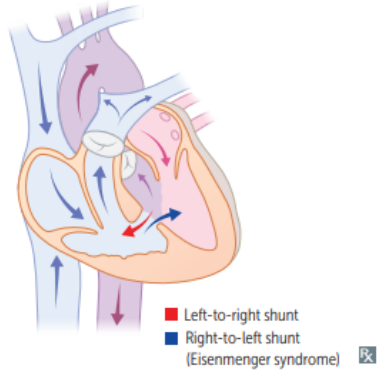
- **Mention two cardiac abnormalities that cause early cyanosis (5Ts)**
  - Persistent **T**runcus arteriosus (1 Trunk)
  - **T**ransposition of great arteries (2 switched vessels)
  - **T**ricuspid atresia (TRI = "3")
  - **T**etralogy of Fallot (Tetra = "4")
  - **T**APVR (5 letters)
- **How to differentiate between respiratory and cardiac hypoxia ?**
  - Hyperoxia test; It is performed by measuring the arterial blood gases of the patient while they breathe room air, then re-measuring the blood gases after the patient has breathed 100% oxygen for 10 minutes

Eisenmenger syndrome

- This image represent a case of untreated VSD. Now, she has developed this complication.

- **What is the name of this complication ?**
  - Central cyanosis due to Eisenmenger syndrome
    - N.B. Answering: “Central cyanosis” alone is not enough
- **Mention two signs that you will see in the hands of this child?**
  - Peripheral cyanosis
  - Finger clubbing

Eisenmenger syndrome	Uncorrected left-to-right shunt (VSD, ASD, PDA) → ↑ pulmonary blood flow → pathologic remodeling of vasculature → pulmonary arterial hypertension. RVH occurs to compensate → shunt becomes right to left when RV > LV pressure (see illustration). Causes late cyanosis, clubbing, and polycythemia. Age of onset varies depending on size and severity of initial left-to-right shunt.



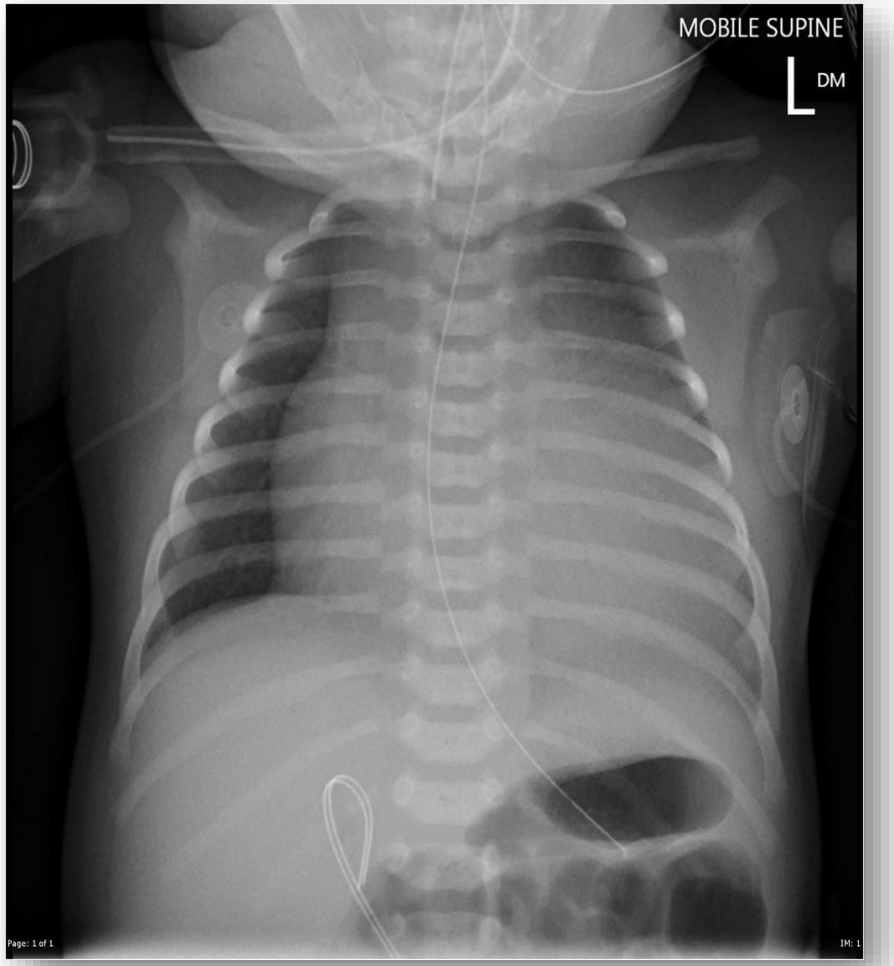
Clubbing

- **Mention 2 clinical findings in this pt.**
  - Hand Clubbing, Cyanosis.
- **Mention 2 diseases cause it.**
  - **C**: **C**yanotic heart disease
  - **L**: **L**ung diseases (e.g., Abscess, Bronchiectasis, Cancer)
  - **U**: **U**lcerative colitis (Inflammatory bowel disease)
  - **B**: **B**enign mesothelioma; **B**irth defects
  - **B**: primary **B**iliary cirrhosis
  - **I** : **I**nfective endocarditis
  - **N**: **N**eurogenic tumor
  - **G**: **G**I malabsorption (Celiac disease)
- **What system do you want to examine for this patient ?**
  - CVS & RS (He have clubbing & cyanosis not only clubbing)

Acyanotic 3 months old

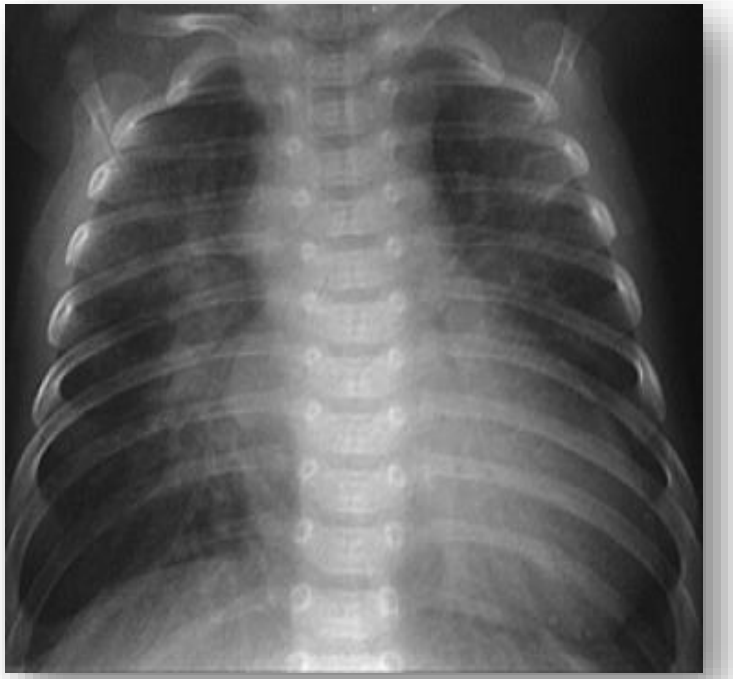
- 3-month-old infant came to the ER with shortness of breath, tachypnea, and failure to thrive.

- **What is the finding in this chest x-ray ?**
  - Cardiomegaly
- **What is the most likely diagnosis ?**
  - Acyanotic congenital heart disease (VSD)



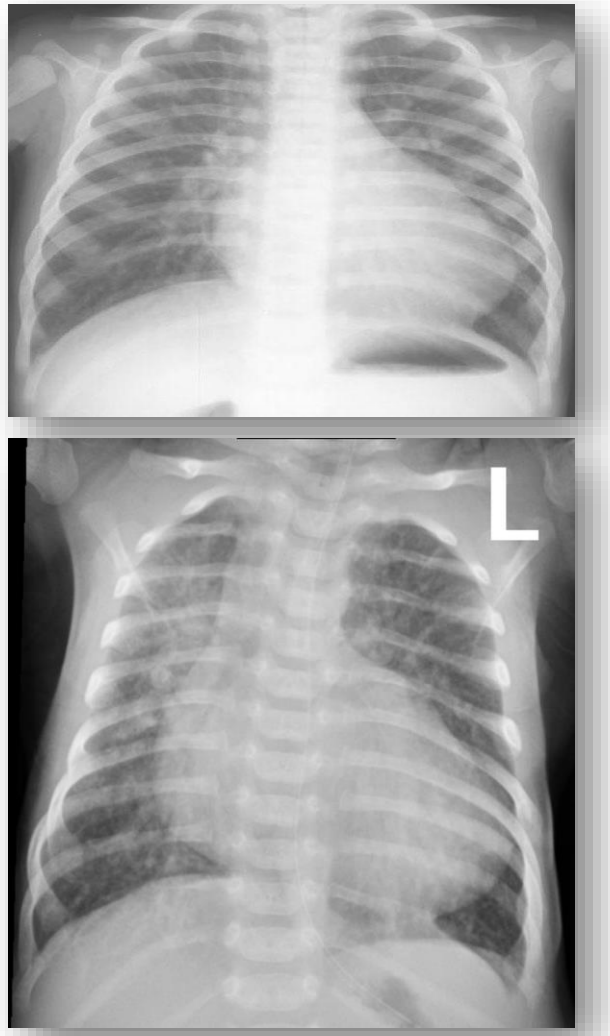
3 weeks old neonate patient presented with cyanosis

- **What is the finding in this CXR ?**
  - Cardiomegaly
- **Mention 2 possible heart abnormalities ?**
  - Tricuspid atresia
  - TAPVR



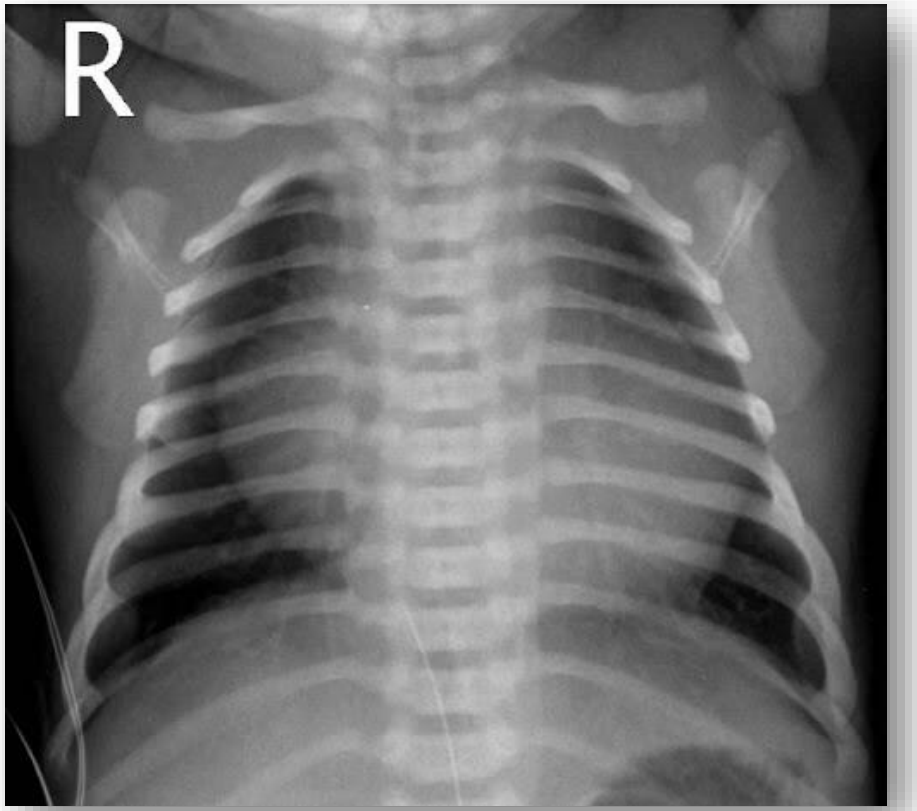
Transposition of great arteries

- **What's the CXR finding ?**
  - Egg-on-string
- **What's the cause of his cyanosis ?**
  - Transposition of great arteries (Two parallel circuits)
- **Management**
  - When transposition is suspected, an infusion of prostaglandin E1 should be initiated immediately
  - Surgical correction is the treatment of choice for neonates with d-TGA and is usually performed within the first 2 weeks of life.



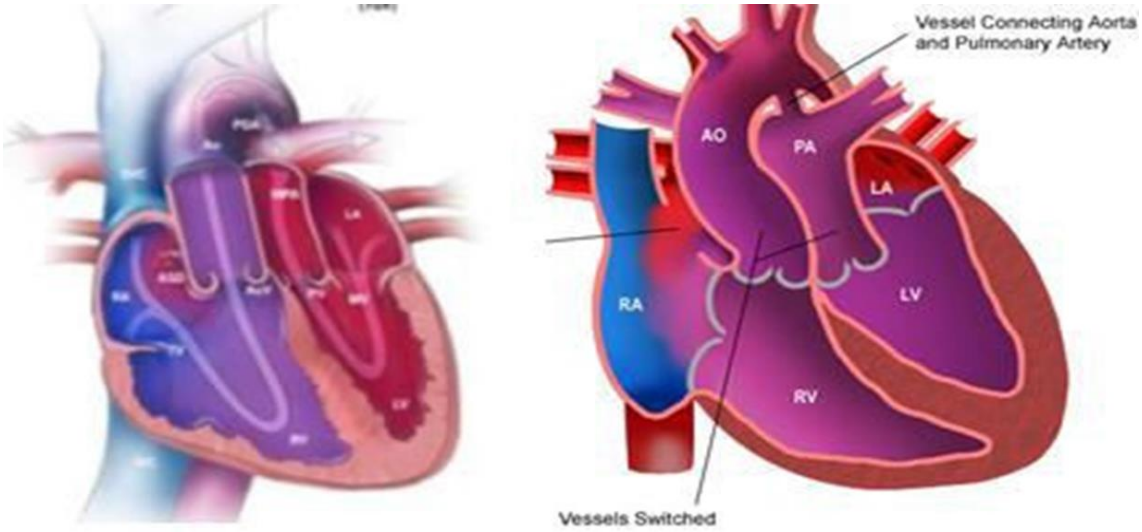
Infant came to you with cyanosis

- **Mention 2 findings on the x ray**
  - Egg-on-string
  - Widening of the mediastinum
- **What's your diagnosis**
  - Transposition of great arteries



Transposition of great arteries

- **What's your Dx. ?**
  - Transposition of great vessels.
- **What's the most common presentation in neonates ?**
  - Central cyanosis.
- **What do you give immediately after birth ?**
  - Prostaglandin (PG E1).

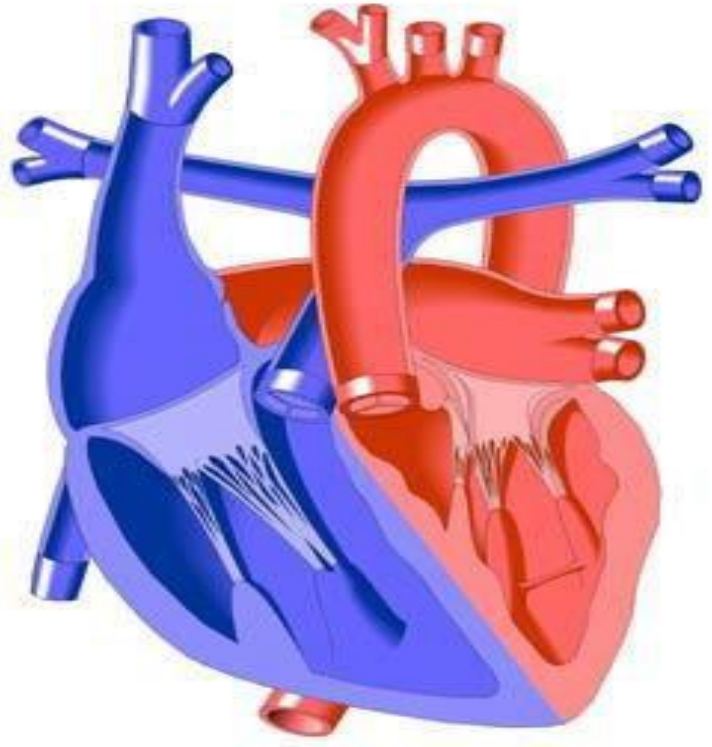


Transposition of great arteries

- 1-day-old neonate, cyanosed with O_2 sat of 75% and PaO_2 = 85 mmHg

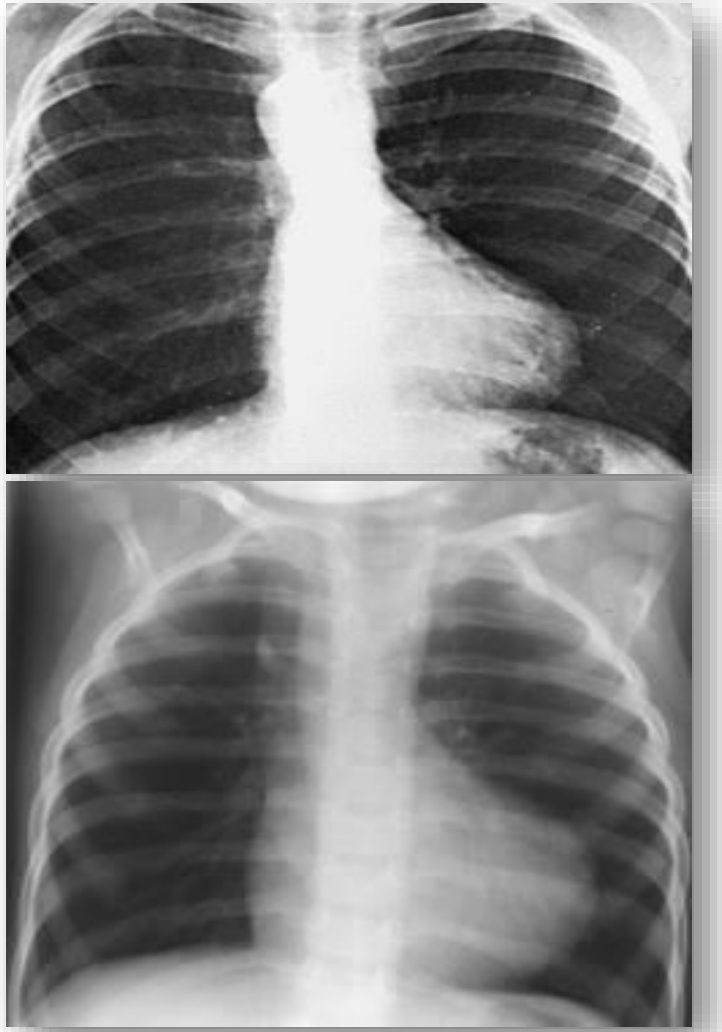
- **What's the CXR finding ?**
  - Egg-on-string.

- **What's the cause of his cyanosis ?**
  - TGA (Two parallel circuits)



6-month-old baby presented with O_2 sat 60%

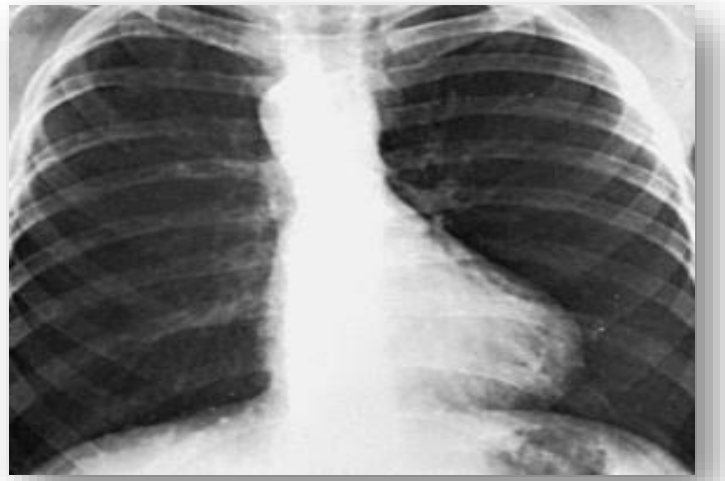
- **Give one finding in the X-Ray ?**
  - Boot shape, hyperlucent lung
- **What is your diagnosis ?**
  - Tetralogy of Fallot
- **Write down 2 modalities of treatment ?**
  - **Surgical**: VSD closure and relieve of RVOT obstruction
    Medical: maintain HCT 45-50%, knee chest position, morphine, phenylephrine, oxygen



Tetralogy of Fallot

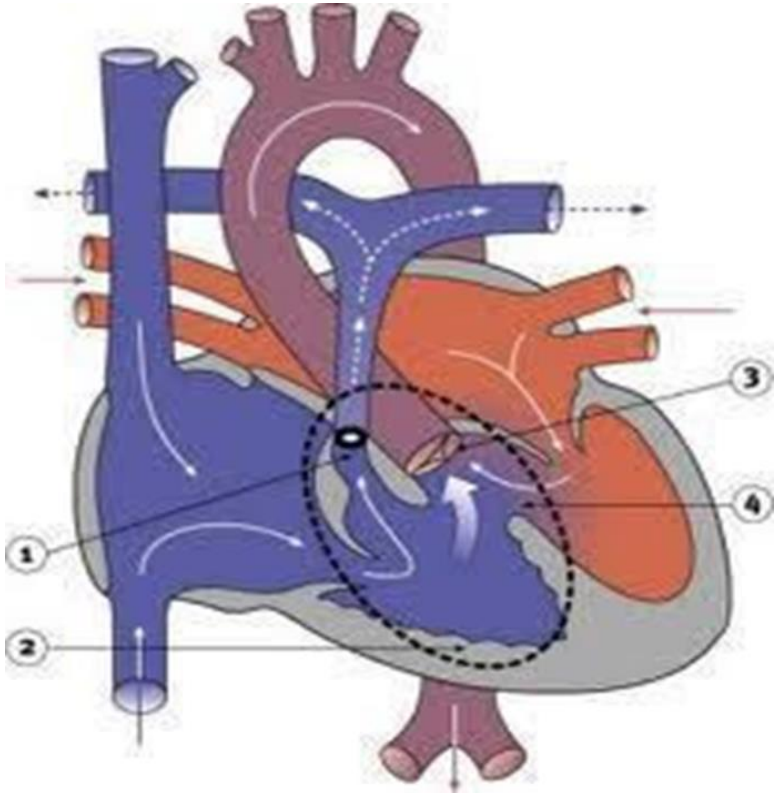
❖A 4 months old boy, has recurrent cyanotic spells with crying at early morning, according to his x-ray which appears above, what is the most likely diagnosis ?

a. Transposition of the great arteries
b. Tricuspid atresia
c. Tetralogy of Fallot
d. Truncus arteriosus
e. Total anomalous pulmonary venous return



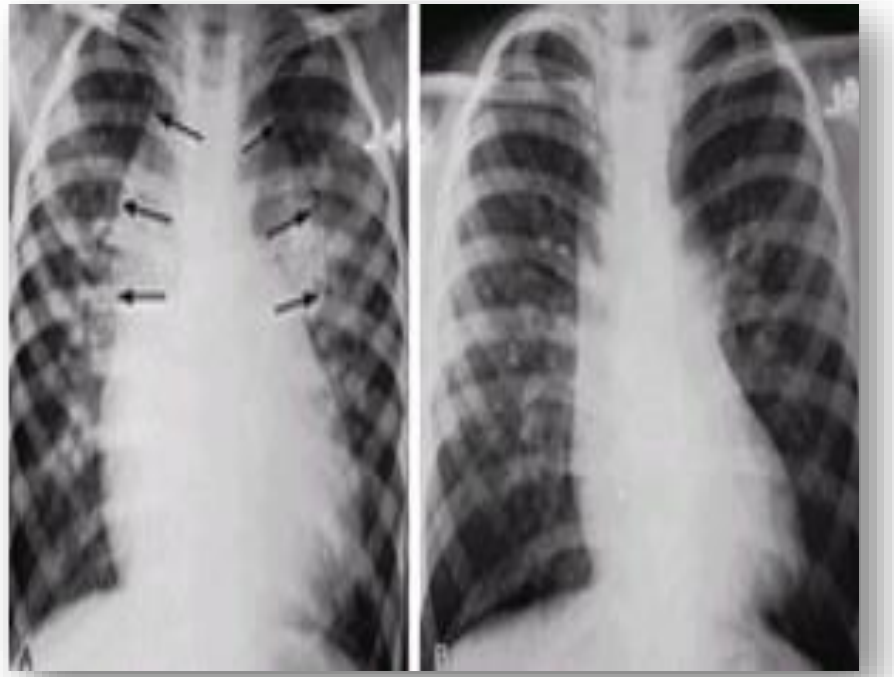
Tetralogy of Fallot

- **What is the disease ?**
  - TOF.
- **Give 3 findings of CVS physical exam.**
  - Ejection systolic murmur.
  - Thrills.
  - Single S2.
  - Cyanosis.
- **Give 2 complications.**
  - Clubbing, Failure to thrive



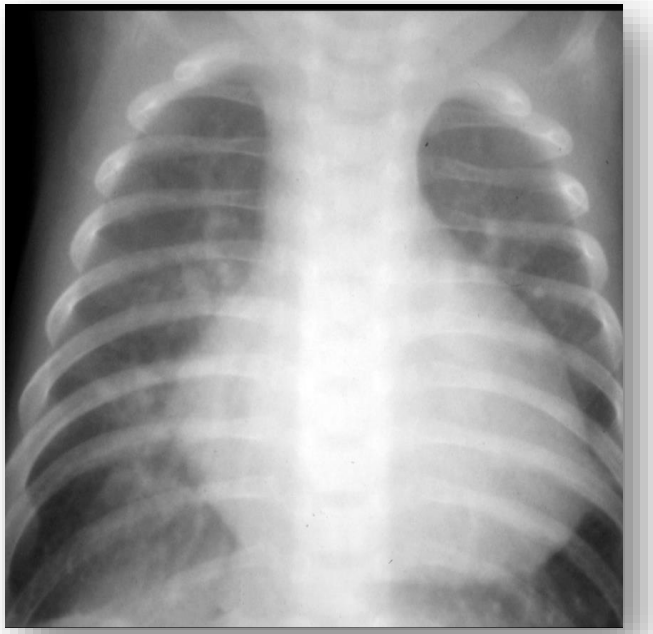
Total Anomalous Pulmonary Venous Return

- **Mention 2 findings**
  - Snowman in a snowstorm (cardiomegaly, pulmonary congestion)
- **What is your diagnosis ?**
  - Total Anomalous Pulmonary Venous Return
- **Management**
  - Initial postnatal management: stabilization of cardiorespiratory status until surgery is performed
  - Surgical repair: recommended in all patients regardless of the severity of disease



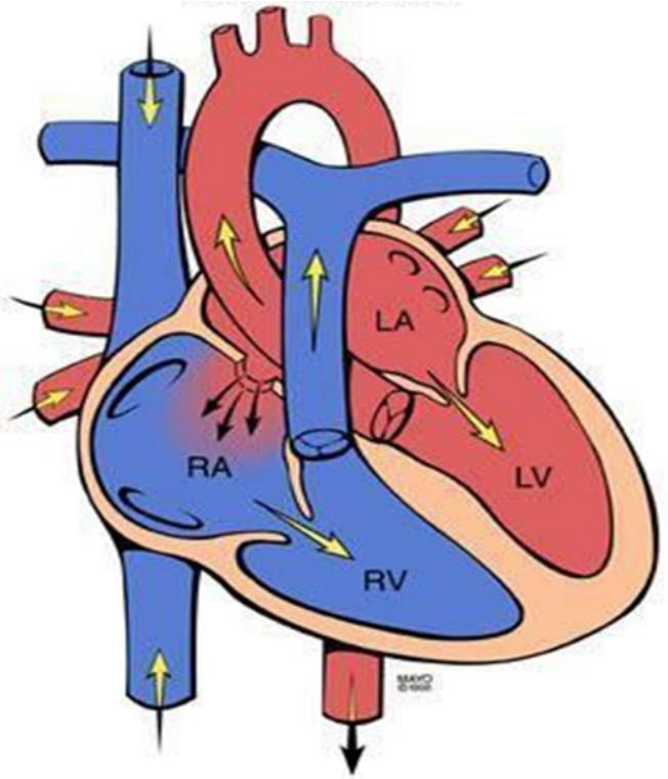
Patient present with holosystolic murmur

- **What is the most common cardiac defect that causes this condition ?**
  - Ventricular septal defect (VSD)
- **If the patient is trisomy 21 what is your Dx ?**
  - Endocardial cushion (AVSD)
- **Write one indication for surgery**
  - Eisenmenger syndrome (reverse of the shunt to be right to left shunt) / congestive heart failure



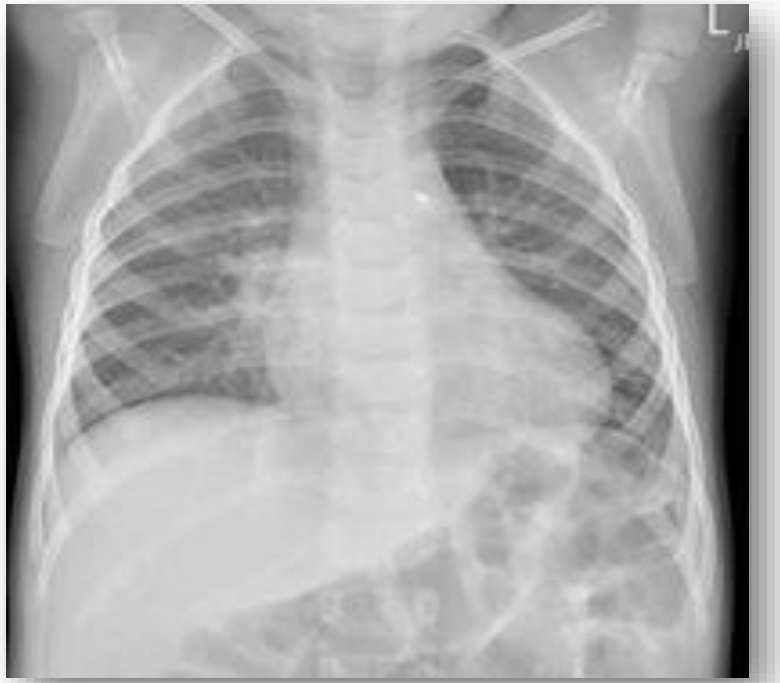
4-year-old healthy boy

- **What is this ?**
  - ASD
- **Give 2 findings upon physical examination ?**
  - Ejection Systolic murmur
  - Fixed splitted S2



2 weeks old baby boy comes with machinery murmur

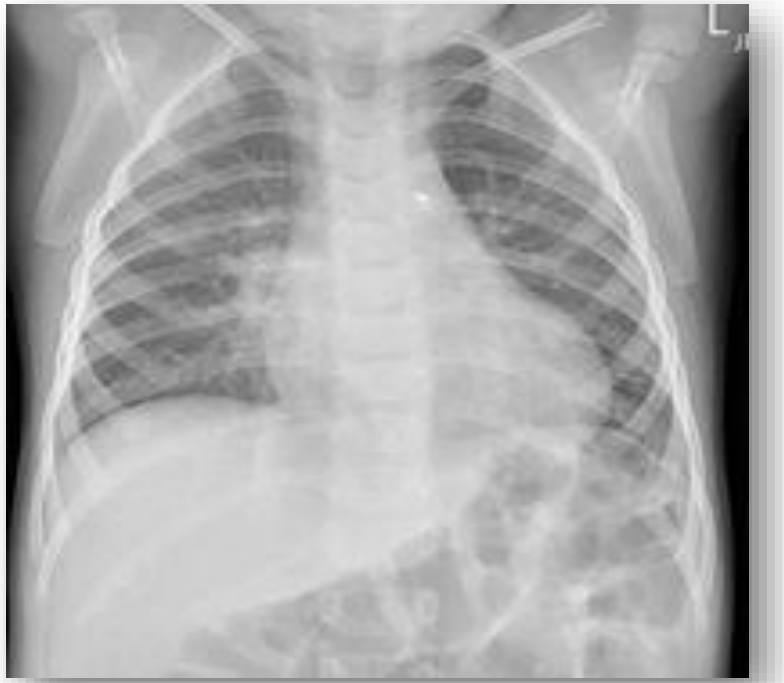
- **What is the diagnosis ?**
  - PDA
- **Give one risk factor**
  - Preterm
- **Treatment**
  - indomethacin or ibuprofen



Patent Ductus Arteriosus

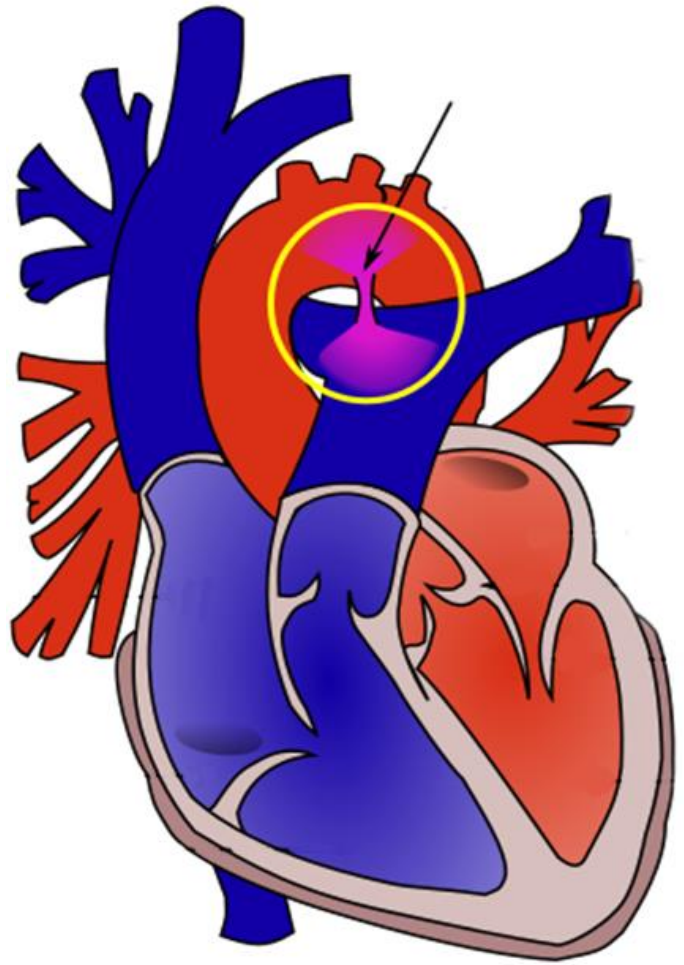
- 3-month-old baby presented with tachypnea, and failure to gain weight, a continuous murmur is heard.

- **What is the most likely diagnosis ?**
  - PDA



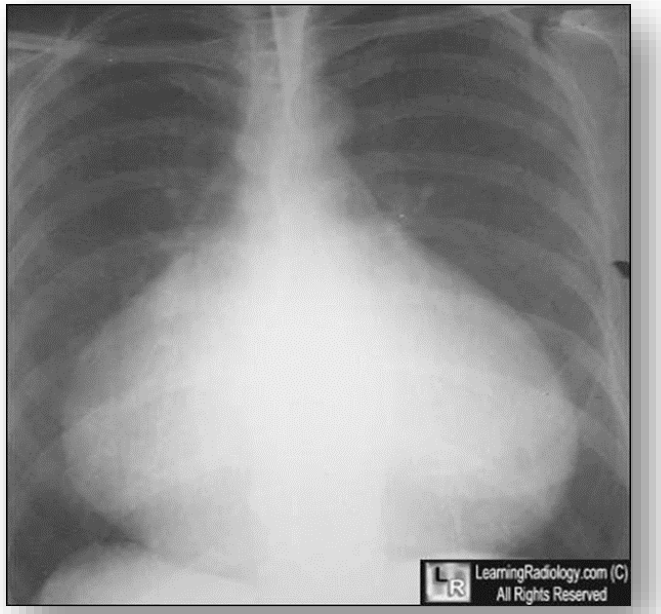
Patent Ductus Arteriosus

- **What is the cardiac anomaly?**
  - PDA
- **Mention 2 signs on physical exam.**
  - Hypertension.
  - Radio-femoral delay.
  - Machinery murmur at infra-clavicular area.
  - Bounding pulse.
- **Give one therapeutic intervention ?**
  - Indomethacin or ibuprofen
  - Surgical ligation of PDA



Pericardial effusion

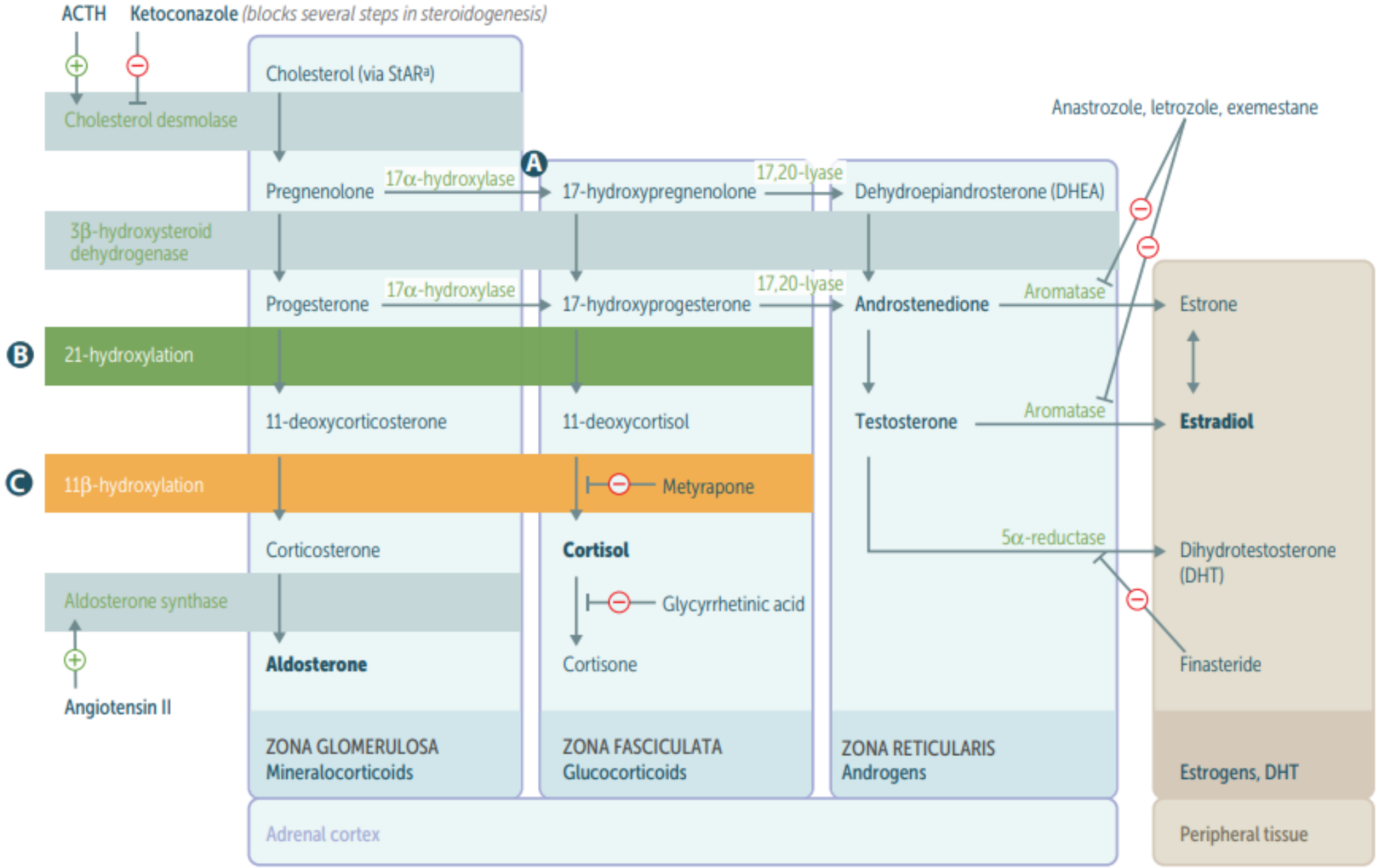
- A 6 YO child, previously healthy, started to complain from fever, SOB, shoulder pain 6 days ago
- **What's the Dx?**
  - Pericardial effusion
- **Mention 2 things you'll hear by auscultation**
  - Friction rub, Muffled heart sounds
  - Others: distended neck veins, hypotension



Congenital Adrenal Hyperplasia

Medium-Yield

Adrenal steroids and congenital adrenal hyperplasias



[a]Rate-limiting step.

شرح

Congenital Adrenal Hyperplasias

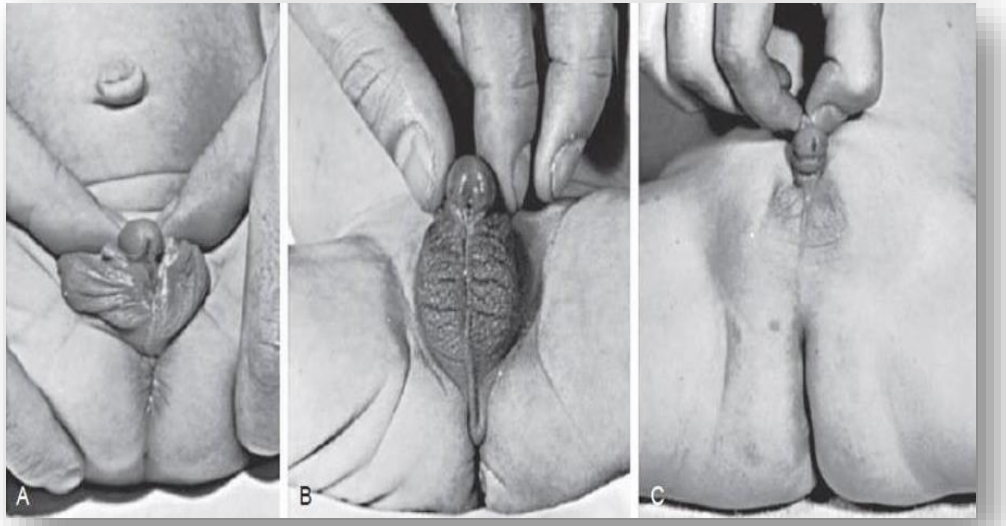
ENZYME DEFICIENCY	MINERALOCORTICOIDS	[K^+]	BP	CORTISOL	SEX HORMONES	LABS	PRESENTATION
A 17α-hydroxylase[a]	↑	↓	↑	↓	↓	↓ androstenedione	XY: atypical genitalia, undescended testes XX: lacks 2° sexual development
B 21-hydroxylase[a]	↓	↑	↓	↓	↑	↑ renin activity ↑ 17-hydroxy-progesterone	Most common Presents in infancy (salt wasting) or childhood (precocious puberty) XX: virilization
C 11β-hydroxylase[a]	↓ aldosterone ↑ 11-deoxycorti-costerone (results in ↑ BP)	↓	↑	↓	↑	↓ renin activity	Presents in infancy (severe hypertension) or childhood (precocious puberty) XX: virilization

[a]All congenital adrenal enzyme deficiencies are autosomal recessive disorders and most are characterized by skin hyperpigmentation (due to ↑ MSH production, which is coproduced and secreted with ACTH) and bilateral adrenal gland enlargement (due to ↑ ACTH stimulation).

If deficient enzyme starts with 1, it causes hypertension; if deficient enzyme ends with 1, it causes virilization in females.

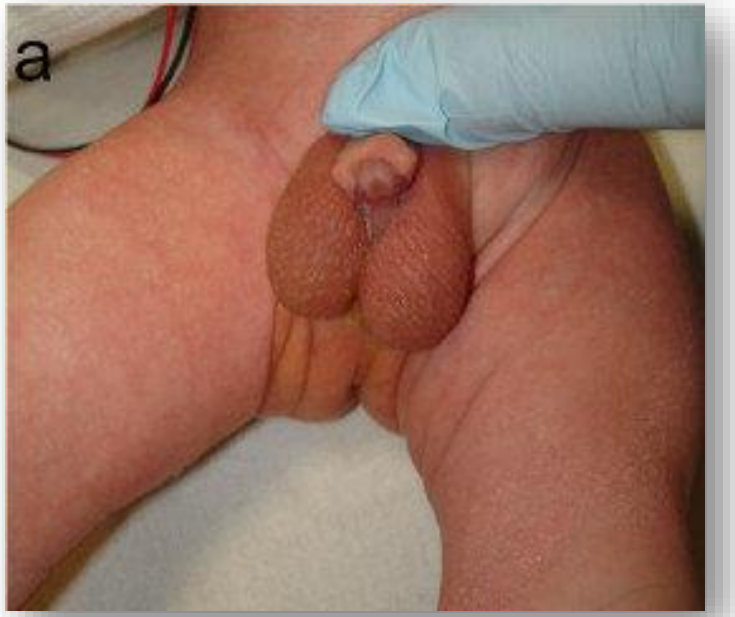
Female patient with ambiguous genitalia

- **What is the diagnosis ?**
  - 21-α hydroxylase or 11β-hydroxylase deficiency
- **Investigation to confirm your diagnosis ?**
  - Measure 17-OH progesterone, 11-Deoxycorticosterone, Corticosterone, Na, K, ACTH, ABG



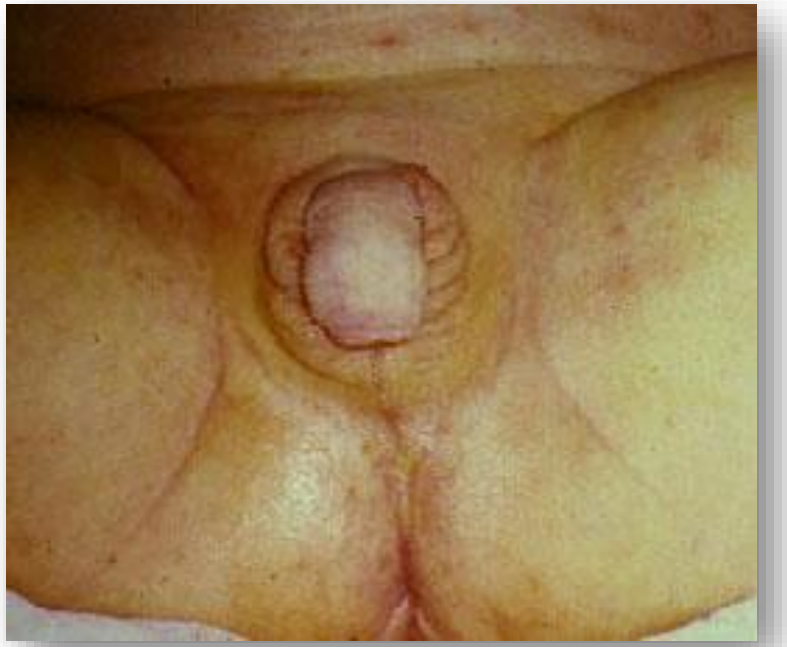
Male patient with ambiguous genitalia

- **What is the deficient enzyme ?**
  - 17 hydroxylase or 3β-hydroxysteroid dehydrogenase deficiency
- **What is the classical presentation for congenital adrenal hyperplasia?**
  - Classically it is due to 21-α hydroxylase:
    - N/V, hypovolemia, hyponatremia, hyperkalemia, hypoglycemia within 7-14 days of life, virilization
- **Investigation to confirm your diagnosis ?**
  - Measure 17-OH progesterone, 11-Deoxycorticosterone, Corticosterone, Na, K, ACTH, ABG
- **What is the best long-term treatment of this patient hyperkalemia ?**
  - Fludrocortisone



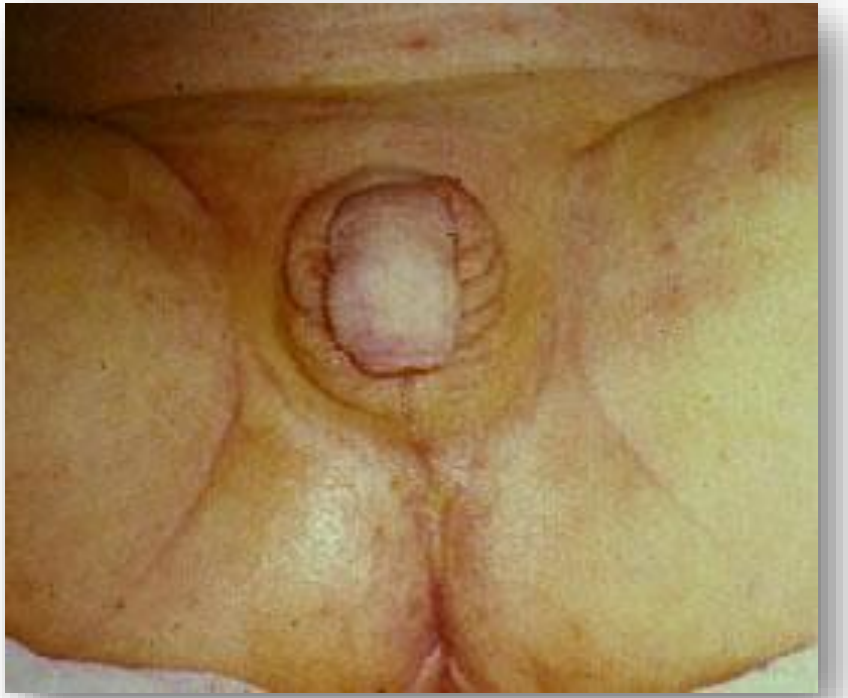
Ambiguous genitalia

- 2 weeks old female (xx) comes to the ER, hypoactive
- **What is th enzyme that is deficient ?**
  - 21-α hydroxylase or 11β-hydroxylase deficiency
- **What are the abnormal electrolytes ?**
  - 21-α hydroxylase deficiency: Hyperkalemia, hyponatremia, hypoglycemia
  - 11β-hydroxylase deficiency: Hypokalemia, hypernatremia, hypoglycemia
- **What is the mode of inheritance?**
  - Autosomal recessive



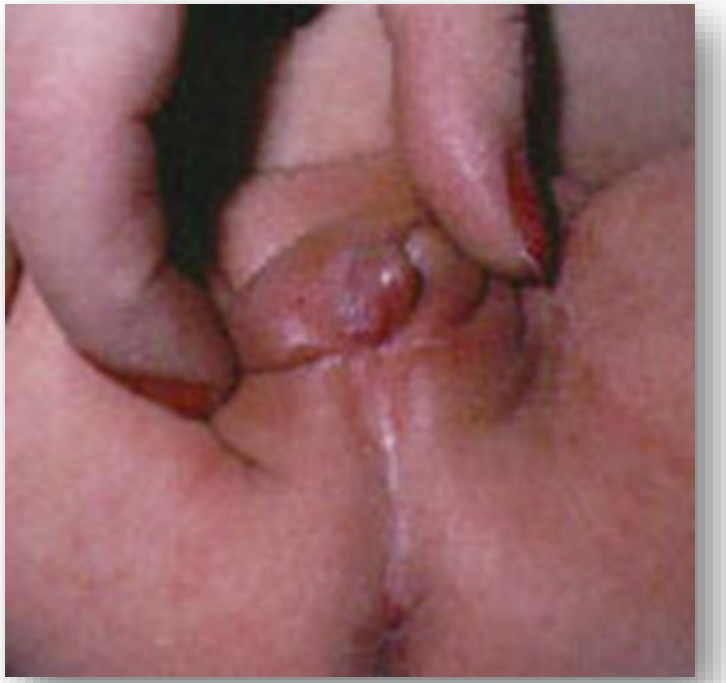
Ambiguous genitalia

- **Female presented with high 17-hydroxyprogesterone, give two presentation**
  - Classic presentation (Salt wasting, absent enzyme): N/V, hypovolemia, hyponatremia, hyperkalemia, hypoglycemia within 7-14 days of life, ambiguous genitalia and virilization
  - Non-classical presentation (Milder form, low enzyme activity): Normal genitalia and as the patient get older, she may experience hirsutism, male pattern baldness, irregular menstruation, and decreased fertility.



Ambiguous genitalia

- Scenario of 2-week-old female patient. with vomiting and diarrhea
- **What is your diagnosis ?**
  - Congenital adrenal hyperplasia
- **What is the investigations ?**
  - Measure 17-OH progesterone, 11-Deoxycorticosterone, Corticosterone, Na, K, ACTH, ABG
- **What is your management ?**
  - Life-long Glucocorticoids
- **Most likely Causative enzyme deficiency ?**
  - 21-α hydroxylase
- **What causes of his seizure ?**
  - Hyponatremia, Hypoglycemia



Ambiguous genitalia

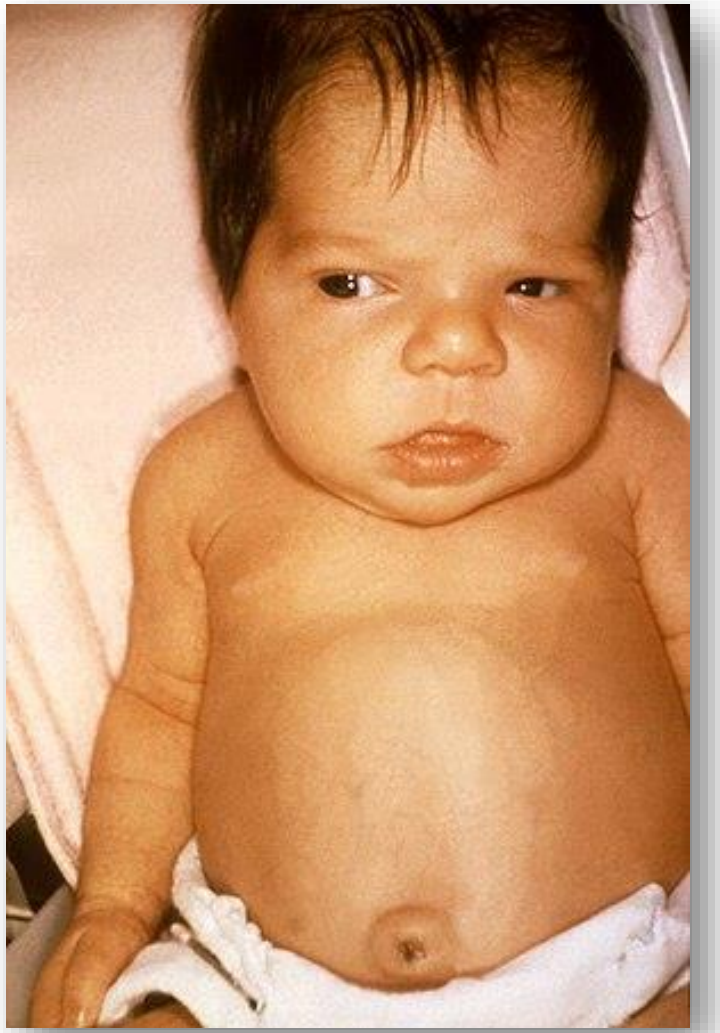
- **Mention 2 signs rather than signs of dehydration**
  - Clitoromegaly, ambiguous genitalia.
- **Mention one diagnostic test**
  - Measure 17-OH progesterone.

Congenital hypothyroidism

Medium-Yield

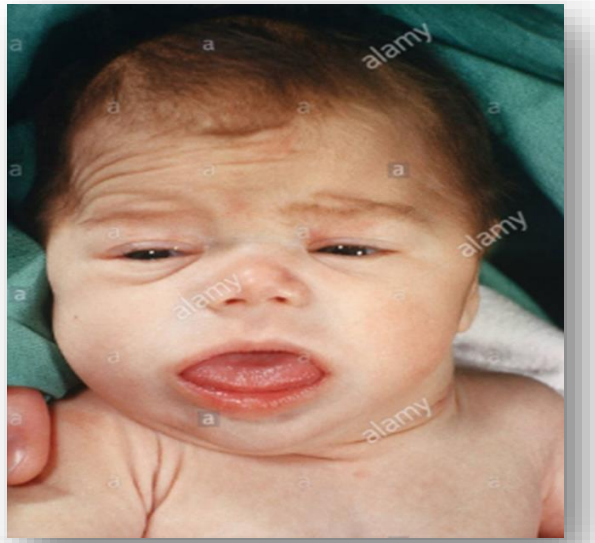
The 7 Ps of congenital hypothyroidism

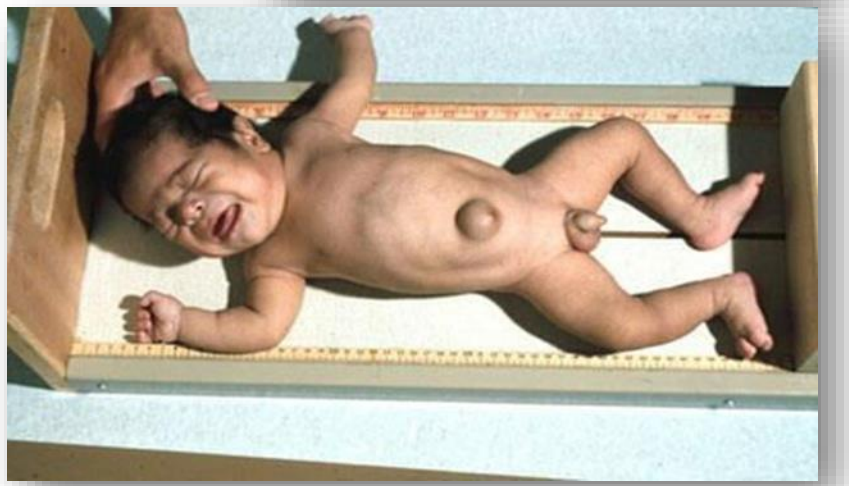
1. **P**rolonged neonatal jaundice
2. **P**ale
3. **P**oor brain development (mental retardation)
4. **P**uffy faced
5. **P**rotuberant tongue (macroglossia) & anterior fontanelle
6. **P**rotruding umbilicus (hernia)
7. **P**ot belly and constipation



Congenital hypothyroidism

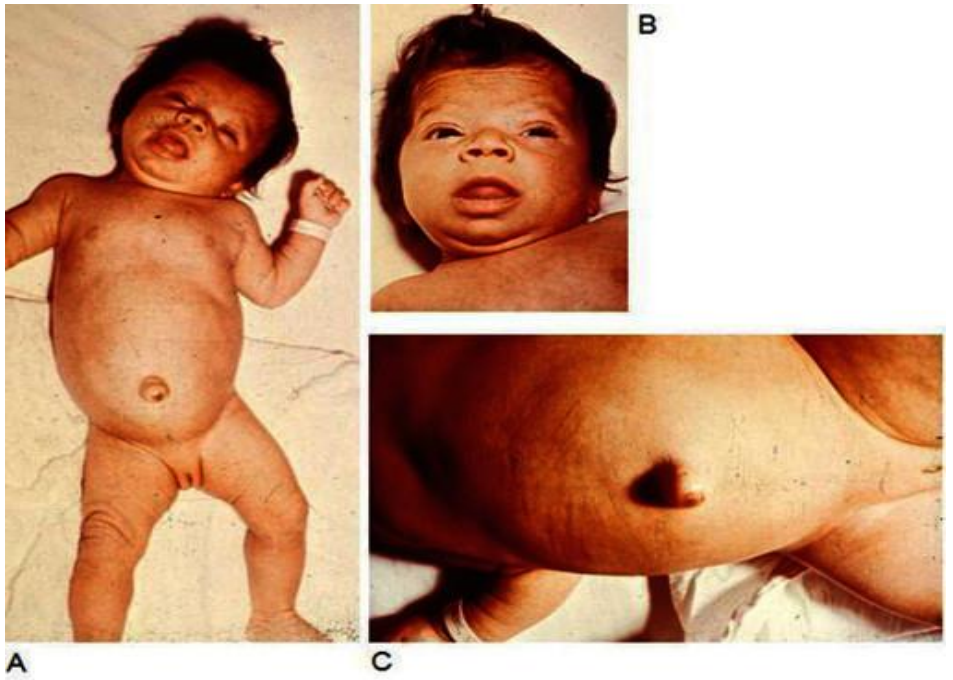
- **What is the diagnosis ?**
  - Congenital hypothyroidism
- **What investigations you would like to order ?**
  - TFT (serum T 3,T4, TSH)
- **What's the expected result ?**
  - High TSH & low T3 & T4.
- **What is your treatment ?**
  - Levothyroxine





Congenital hypothyroidism

- **Mention 2 signs.**
  - Macroglossia, Umbilical hernia.
- **What's your Dx.?**
  - Congenital Hypothyroidism.
- **What lab test you want to do?**
  - TFT (serum T 3,T4, TSH).
- **What's the expected result?**
  - High TSH & low T3 & T4.

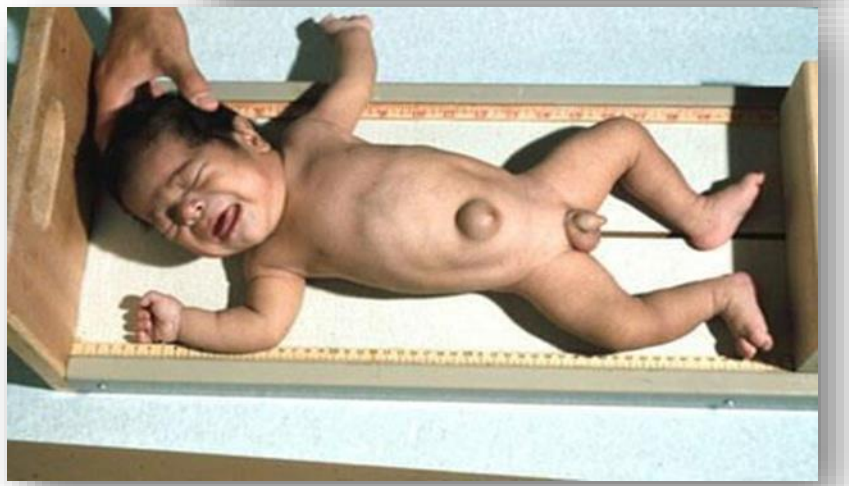


Congenital hypothyroidism

- 1-month-old infant presented to you with a history of week cry and hypoactivity since birth

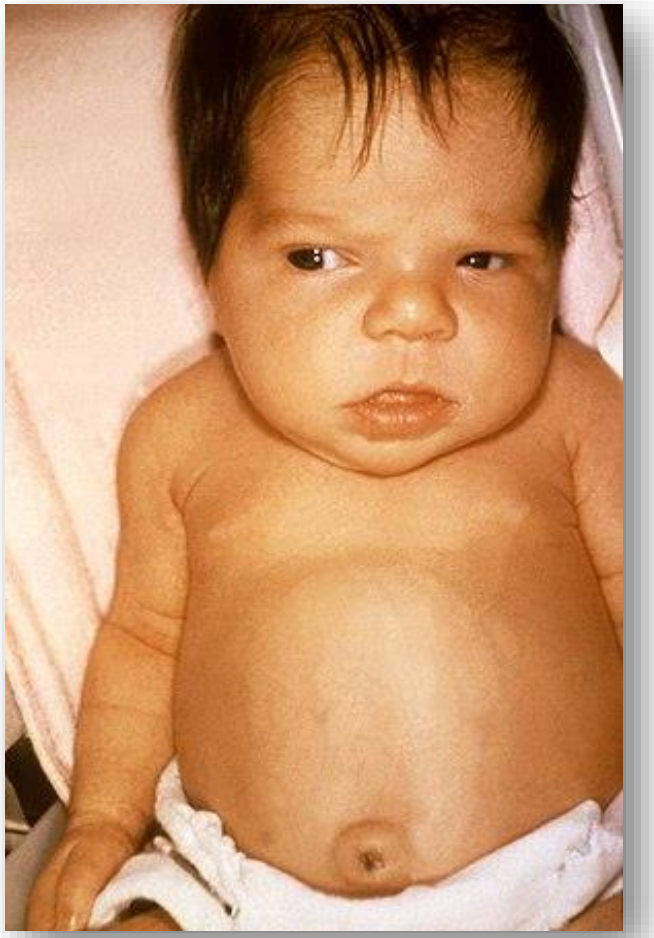
- **Name 3 signs**
  - Depressed nasal bridge
  - Macroglossia
  - Umbilical hernia
  - Open anterior and posterior fontanelle

- **What is the most likely Diagnosis ?**
  - Congenital hypothyroidism



Congenital hypothyroidism

- **What is the cause of this condition ?**
  - Congenital hypothyroidism
- **Write 2 physical findings**
  - Depressed nasal bridge, Macroglossia, Umbilical hernia, Open anterior and posterior fontanelle
- **What is your treatment ?**
  - Levothyroxine
- **If he have goiter what the cause of his disease ?**
  - Dyshormonogenesis, endemic iodine deficiency or maternal antithyroid drug



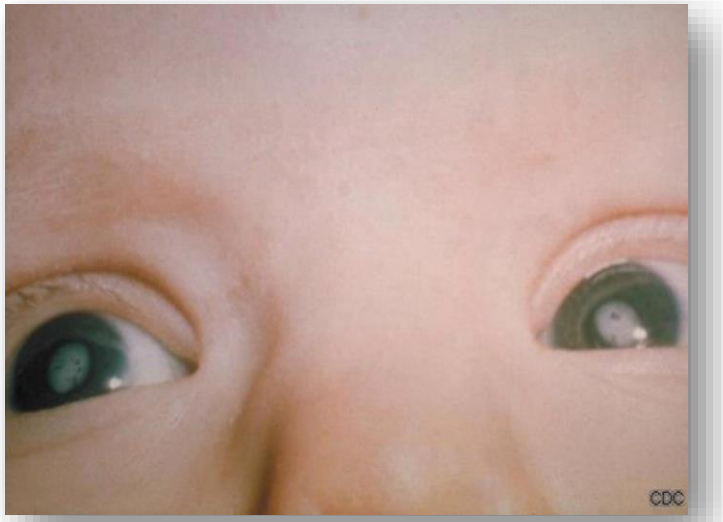
Congenital cataracts

Low-Yield

Congenital cataracts

1. Hereditary congenital cataracts
2. Caused by TORCH infections (esp., rubella)
3. Associated with the following comorbidities/syndromes:
   A. Galactosemia
   B. Tetany (hypocalcemia)
   C. Myotonic dystrophy
   D. Skin diseases
   E. Trisomy 21 (in early childhood)
   F. Trisomy 13
   G. Trisomy 18
   H. Alport syndrome
   I. Neurofibromatosis type 2
   J. Galactokinase deficiency
   K. Marfan syndrome

Cataracts



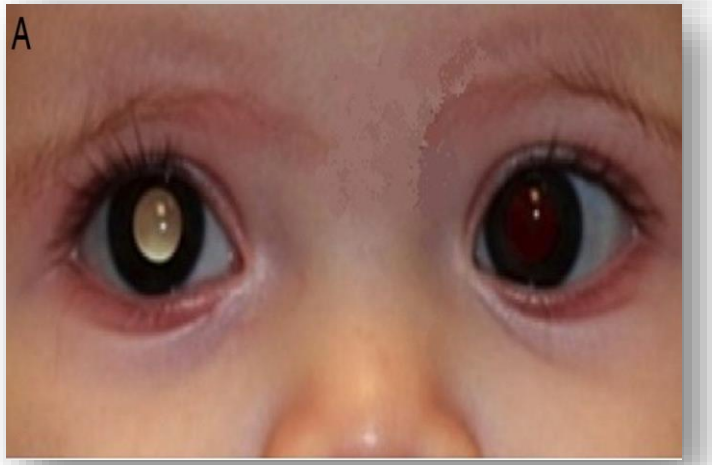
- **What's your Dx.?**
  - Congenital Cataract.
- **Mention 2 causes.**
  - Rubella, Galactosemia.

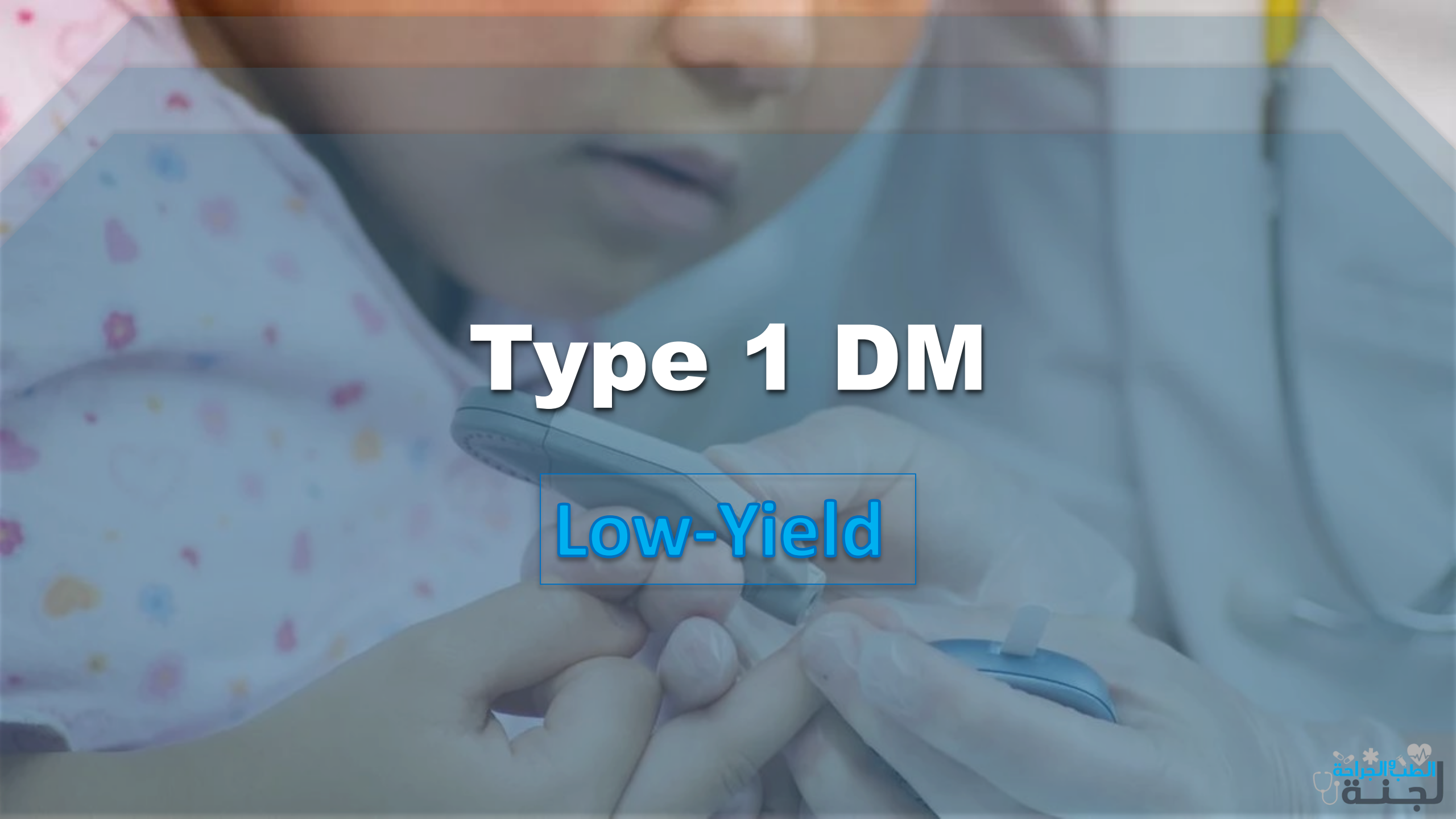
سؤال سادسة كان فيه وصف يدل انه "Galactosemia" لكن نص السؤال مش مكتوب

- **What's your Dx.?**
  - Galactosemia
- **What is the treatment ?**
  - Galactose free diet

Cataracts

- **What is the finding ?**
  - Leukocoria (absent red reflex / white reflex)
- **Mention two causes of such condition ?**
  - Cataracts (congenital rubella , galactosemia)
  - Retinoblastoma
  - Retinopathy of prematurity





Type 1 DM

Low-Yield

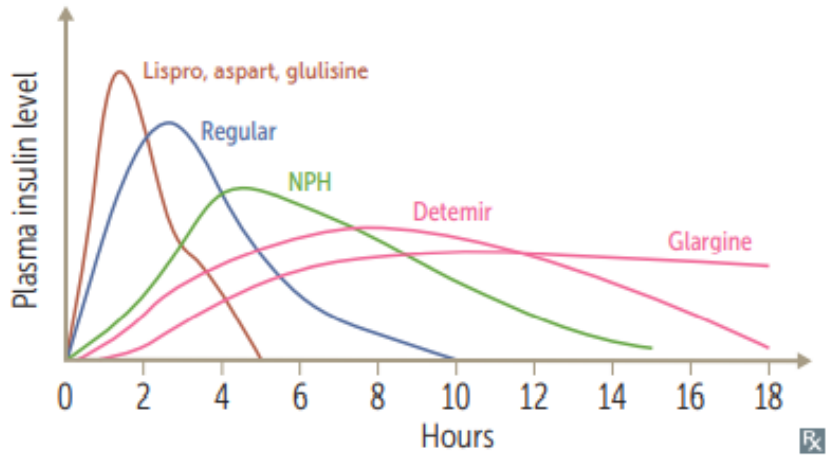
Type 1 DM

- **What is the type of insulin ?**
  - Long acting
- **What is the duration of action ?**
  - 24 hours
- **Mention 4 diagnostic criteria for type 1 DM**
  - Symptoms of DM + random casual plasma glucose >200 mg/dL
  - Fasting plasma glucose > 126 mg/dL
  - 2hr plasma glucose during the OGTT > 200 mg/dL
  - HbA_{1c} > 6.5

إضافي

Insulin preparations

DRUG CLASS	MECHANISM	ADVERSE EFFECTS
Insulin preparations		
Rapid acting (1-hr peak): Lispro, Aspart, Glulisine (no LAG) Short acting (2–3 hr peak): regular Intermediate acting (4–10 hr peak): NPH Long acting (no real peak): detemir, glargine	Bind insulin receptor (tyrosine kinase activity) Liver: ↑ glucose storage as glycogen Muscle: ↑ glycogen, protein synthesis Fat: ↑ TG storage Cell membrane: ↑ K^+ uptake	Hypoglycemia, lipodystrophy, hypersensitivity reactions (rare), weight gain



- Important life note: All types of insulin can cause hypoglycemia; even insulin glargine, which is long-acting peak less insulin. How can insulin glargine cause hypoglycemia if it peak less you ask ? Simply you rise the baseline insulin in the body and thus increase the baseline uptake of glucose from blood resulting in recurrent hypoglycemic attacks through out the day especially when the patient is fasting i.e., during sleeping ☺

Neonatology

Neonatal Jaundice

Medium-Yield

Neonatal Jaundice

- It is due to increased serum bilirubin, typically visible when > 5 mg/dL
- It is either physiological or pathological
- Most common cause of neonatal jaundice is physiological jaundice

Physiological neonatal jaundice	Pathological neonatal jaundice
Always unconjugated hyperbilirubinemia	Can be either unconjugated or conjugated
> 24h of life	Can occur < 24h of life
< 15 mg/dL	May rise > 15 mg/dL
< 5 mg/dL/day	> 5 mg/dL/day
Pathophysiology 1. Short lifespan of erythrocytes in the newborn 2. Insufficient hepatic bilirubin metabolism 3. ↑ Enterohepatic circulation of bilirubin	1. Increased production of bilirubin 2. Decreased hepatic uptake 3. Decreased conjugation 4. Impaired excretion 5. Increased enterohepatic circulation

Pathological neonatal jaundice

1. **Increased production of bilirubin**
   - Hemolytic disease of the newborn (e.g., ABO incompatibility, Rh incompatibility)
   - Erythrocyte enzyme defects (e.g., G6PD deficiency, pyruvate kinase deficiency)
   - Erythrocyte membrane defects (e.g., hereditary spherocytosis)
   - Hemoglobinopathies (e.g., sickle cell anemia, thalassemia)
   - Hematomas (e.g., vacuum-assisted delivery, vitamin K deficiency bleeding)
   - Infection/sepsis
   - Polycythemia
2. **Decreased hepatic uptake**
   - Rotor syndrome

Pathological neonatal jaundice

3. **Decreased conjugation**
   - Gilbert syndrome
   - Crigler-Najjar syndrome
   - Hypothyroidism
4. **Impaired excretion**
   - TORCH infections
   - Dubin-Johnson syndrome
   - Sepsis
   - Biliary atresia
   - Biliary/choledochal cyst
5. **↑ Enterohepatic circulation of bilirubin**
   - GI obstruction (e.g., pyloric stenosis, bowel obstruction)
   - Cystic fibrosis
   - Breast milk jaundice
   - Breastfeeding jaundice
   - Malnutrition

Breastfeeding & Breast milk jaundice

Breastfeeding jaundice

- **Definition**: a type of neonatal jaundice caused by insufficient breastfeeding
- **Pathophysiology**: insufficient breast milk intake → lack of calories and inadequate quantities of bowel movements to remove bilirubin from the body → ↑ enterohepatic circulation → increased reabsorption of bilirubin from the intestines → unconjugated hyperbilirubinemia
- **Clinical features**: onset within 1 week
- **Treatment**: increase breastfeeding sessions, rehydration

Breast milk jaundice

- **Definition**: a type of neonatal jaundice caused by increased levels of β-glucuronidase in maternal breast milk
- **Pathophysiology**: increased concentration of β-glucuronidase in breast milk → ↑ deconjugation and reabsorption of bilirubin → persistence of physiologic jaundice with unconjugated hyperbilirubinemia
- **Clinical features**: onset within 2 weeks after birth; lasts for 4–13 weeks
- **Treatment**: continued breastfeeding and supplementation with formula feeds

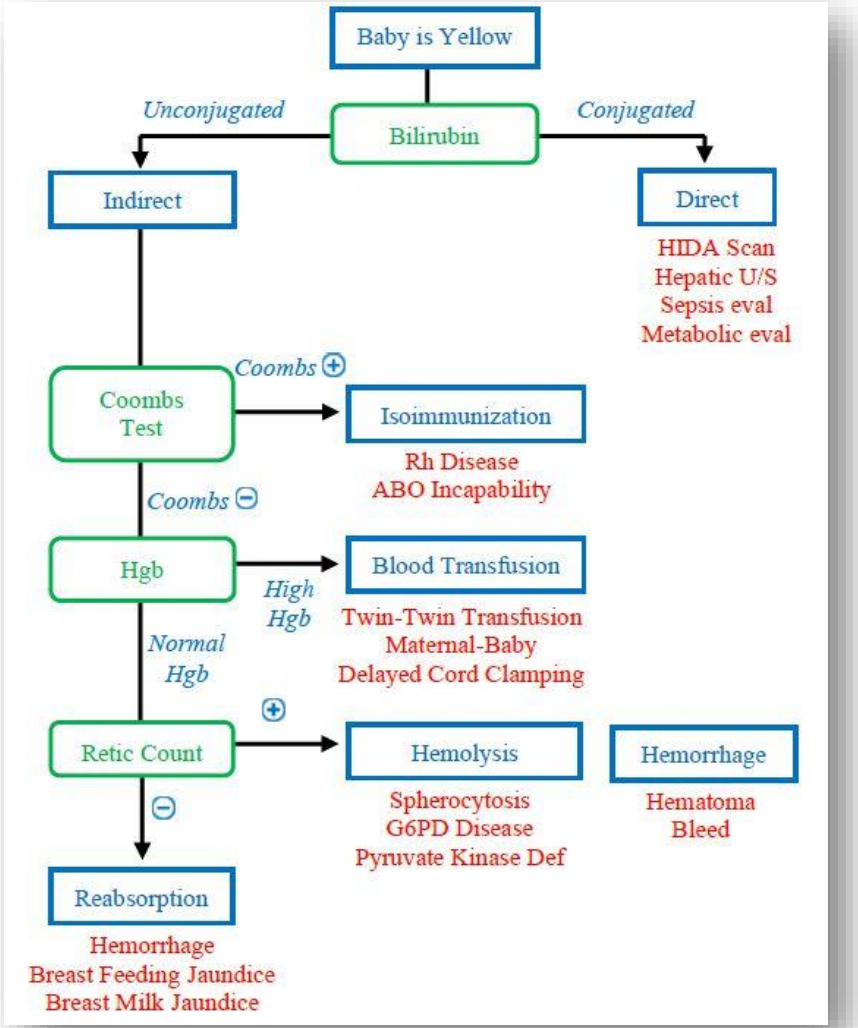
Neonatal Jaundice – Screening

- All neonates should be assessed for jaundice regularly every 8-12h while in the hospital
- When should you start a full work-up ?
  - **Conjugated Hyperbilirubinemia** (conjugated > 20% of TSB); any time
  - **Cord bilirubin** (if obtained) > 70 μmol/L = hemolysis
  - **An increase of** > 85 μmol/L/day or 0.5 μmol/L/hour = hemolysis
  - **Total bilirubin** > 170 μmol/L at or before 24 hours of life
  - **Total bilirubin** > 250 μmol/L in any infant

Neonatal Jaundice – Work-up

1. **Hx and physical**

2. **Labs**
   A. Total and conjugated bilirubin
   B. Mother & infants blood group, Coombs
   C. CBC – Hgb, Hct, Reticulocyte count
   D. Total serum protein and serum albumin (provide information about the nutritional status of a neonate)
   E. Liver enzymes
   F. TSH and free T_4
   G. G6PD activity



Neonatal Jaundice – Treatment

Phototherapy

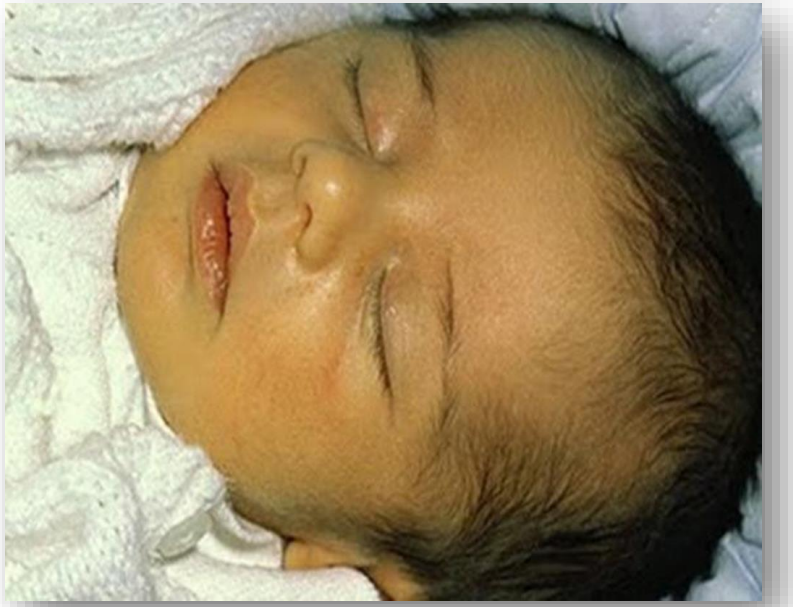
- Primary treatment in neonates with unconjugated hyperbilirubinemia
- For infants born ≥ 35 weeks gestation, threshold bilirubin levels for treatment are based on the American Academy of Pediatrics (AAP) phototherapy nomogram
  - Low-Risk: ≥ 38 weeks and well
  - Medium-Risk: ≥ 38 weeks with risk factors or 35-37 weeks and well
  - High-Risk: 35-37 with risk factors
- Phototherapy is continued until total bilirubin levels < 15 mg/dL

Exchange transfusion

- Most rapid method for lowering serum bilirubin concentrations
- Indications
  - Threshold in a 24-hour-old term baby is a total serum bilirubin value > 20 mg/dL
  - Inadequate response to phototherapy
  - Acute bilirubin encephalopathy
- Exchange blood in quantities of 5–20 mL via an umbilical venous catheter until total serum bilirubin is < 95th percentile on nomogram

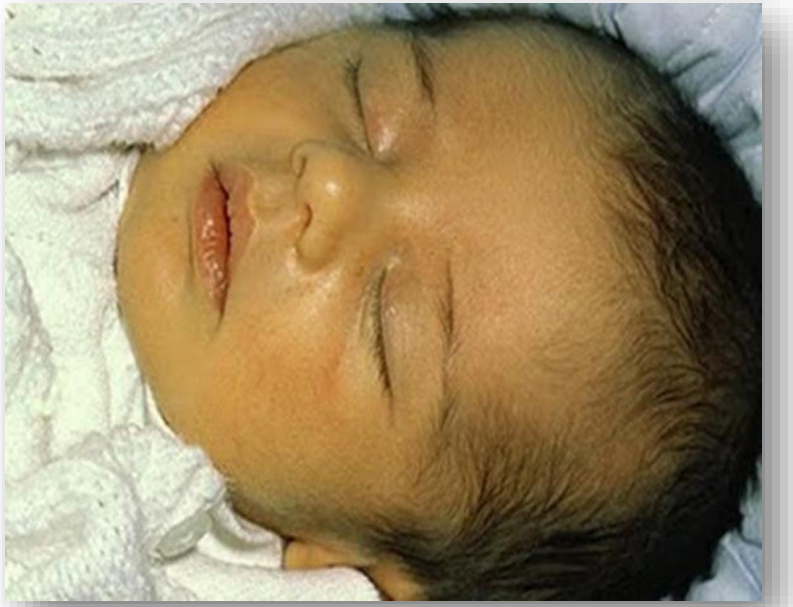
Neonatal Jaundice

- **What is the Diagnosis ?**
  - Neonatal jaundice
- **When is it seen ?**
  - When Bilirubin levels > 5 mg/dL



Neonatal Jaundice

- **Mention 2 causes for this condition seen in 2 months old baby, who has elevated indirect bilirubin levels**
  - Hemolysis
  - Hematomas
  - Conjugation disorders (crigler-najjar syndrome type 1)



Neonatal Jaundice

- At second day of life of this child he presented with seizure, poor sucking, hypotonia

- **What are the tests you should perform ?**
  - Random blood sugar
  - Serum bilirubin level
  - CBC
  - Na serum levels

- **What is the cause of seizure ?**
  - Kernicterus

A baby in NICU

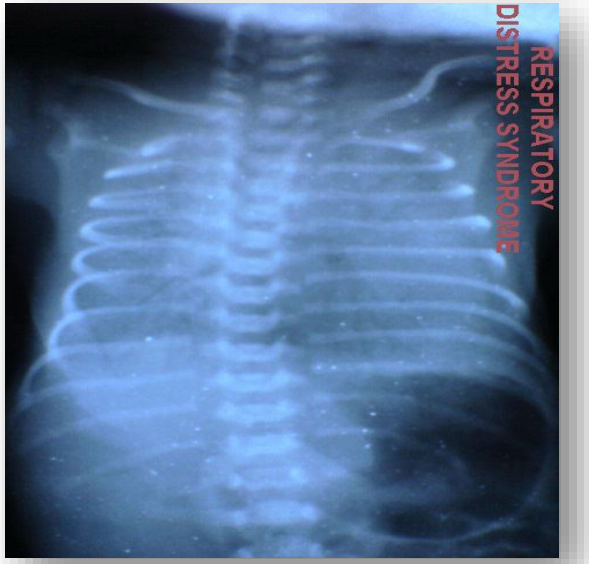
- **What is this type of treatment ?**
  - Phototherapy
- **What is the mechanism of action ?**
  - Transform unconjugated bilirubin to water soluble form to excrete it out of the body in Urine
- **Mention 4 causes for jaundice**
  - G6PD Deficiency
  - Hereditary Spherocytosis
  - Hematoma
  - Crigglar-Najjar Syndrome
  - Any cause of indirect hyperbilirubinemia

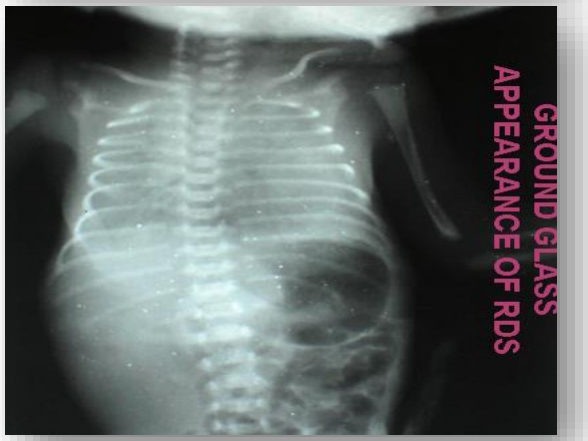
Respiratory

Medium-Yield

Neonatal respiratory distress syndrome

- **Term**: Preterm
- **Risk factors**: Preterm delivery, Male sex, Maternal diabetes
- **Onset of symptoms**: Within the first minutes/hours after birth
- **Clinical features**
  - Tachypnea, Increased breathing effort, Cyanosis, Hypoxia, Decreased breathing sounds
- **Imaging**
  - Diffuse, fine, reticulogranular (ground-glass) densities
  - Low lung volumes and air bronchograms
- **Treatment**
  - Supportive care, Nasal CPAP, Endotracheal administration of artificial surfactant





Neonatal respiratory distress syndrome

➢Premature 34 weeks

❖**What's the name of this sign ?**

- Ground glass appearance and air bronchogram

❖**What's the most likely Dx.?**

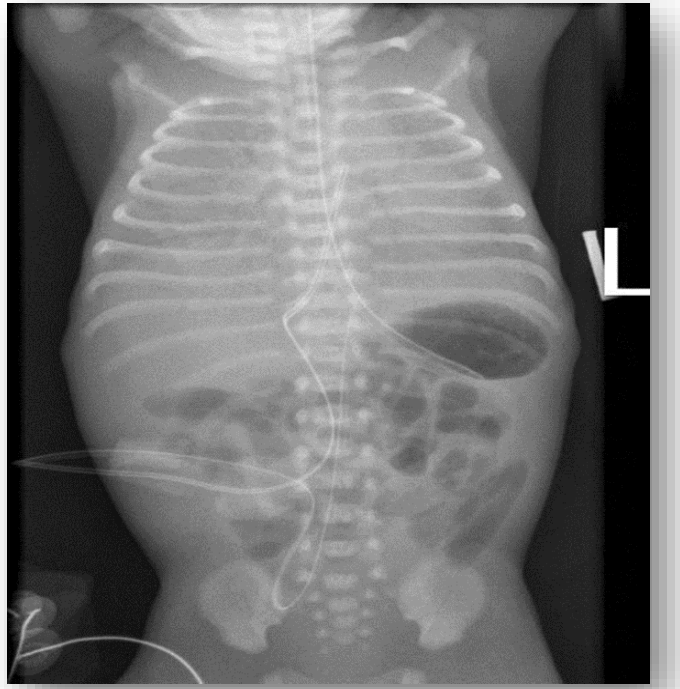
- Respiratory Distress Syndrome (RDS)

❖**What's your management ?**

- Nasal canula if failed nasal CPAP if failed mechanical ventilation with surfactant admission
- Supportive care (antibiotics)

❖**What are the possible complications ?**

- Bronchopulmonary dysplasia
- Pneumothorax
- Patent ductus arteriosus



Indications to give surfactants

- Gestational age < 30 weeks
- Failure of CPAP

Neonatal respiratory distress syndrome

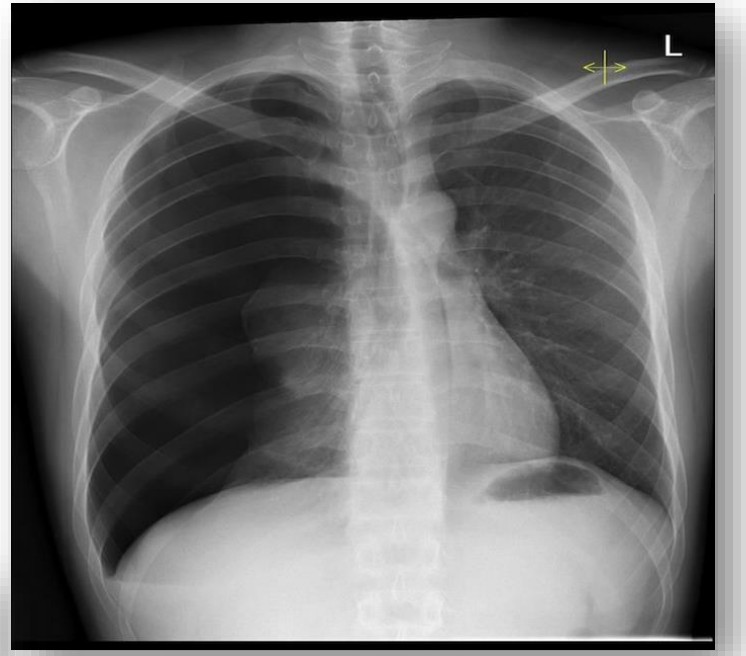
- Premature 28 weeks

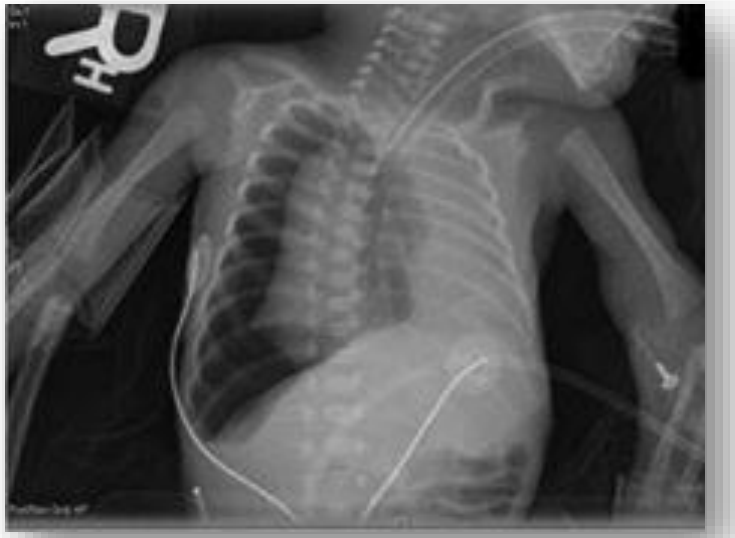
- **What's your management ?**
  - Nasal canula if failed nasal CPAP if failed mechanical ventilation
  - Surfactant admission (must be given gestational age > 30W)
  - Supportive care (antibiotics)

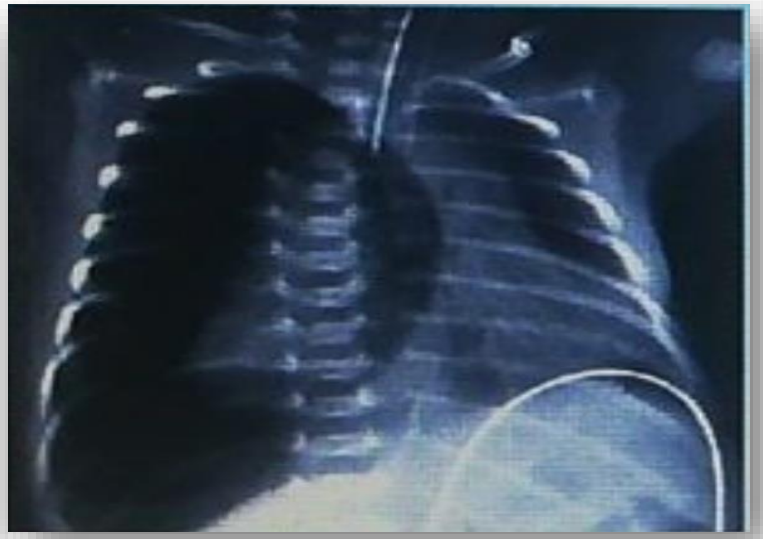
- **What is your O_2 targets ?**
  - SpO_2: 88-92%
  - P_{CO2}: 45-55 mmHg
  - Maintain PaO_2 at 50-80 mmHg

Pneumothorax as complication of intubation in NRDS case

- **What is your diagnosis ?**
  - Tension pneumothorax
- **What is your immediate emergent management ?**
  - Needle decompression / chest tube

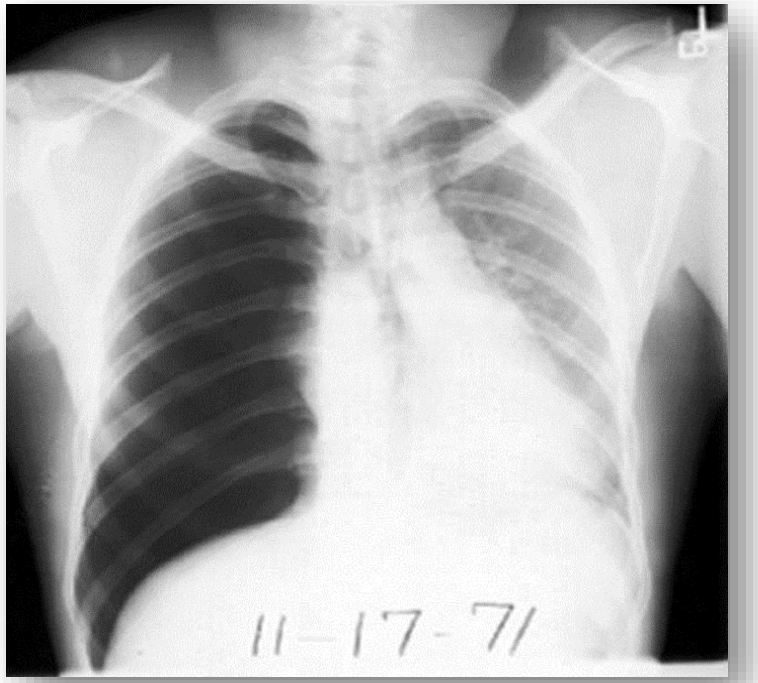






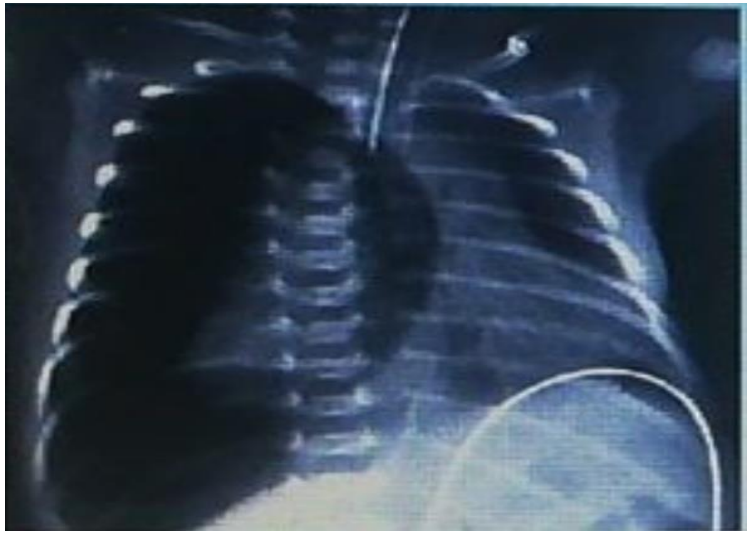
Pneumothorax as complication of intubation in NRDS case

- **What's your Diagnosis ?**
  - Tension Pneumothorax.
- **Give 2 signs on CXR.**
  - Hyper-lucent Rt. lung field
  - Shifted mediastinum (Tracheal deviation to Lt)
  - Heart shadow shifted to Lt
- **What's the treatment ?**
  - Needle Thoracostomy & Chest tube.

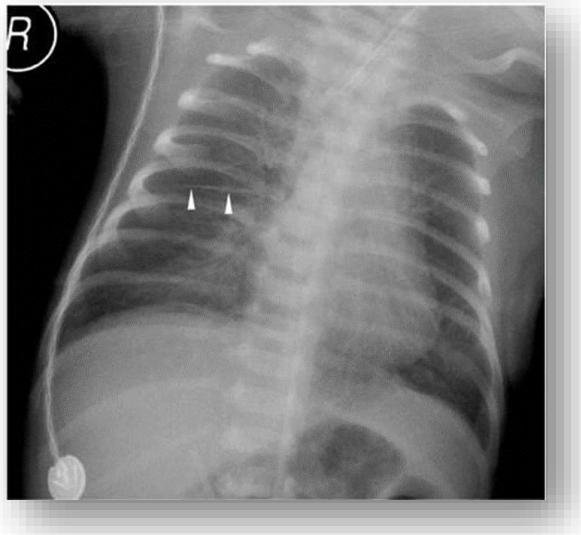


Pneumothorax as complication of intubation in NRDS case

- Newborn male baby, Term = 30 weeks, birth weight 1.2 Kg, had severe RDS, had been connected to mechanical ventilation and given one dose of surfactant, his O2 saturation has been significantly improved after surfactant therapy. Two hours later the baby had sudden onset deterioration with tachycardia, hypertension, severe cyanosis. decreased right side air entry.
- **According to his x-ray which appears in the figure, the treatment of choice is**
  - Start IV Prostaglandin (PGE1)
  - Start IV indomethacin
  - Start IV bolus of normal saline
  - Immediate chest tube insertion on the right side of the chest
  - Another dose of surfactant is mandatory for this situation



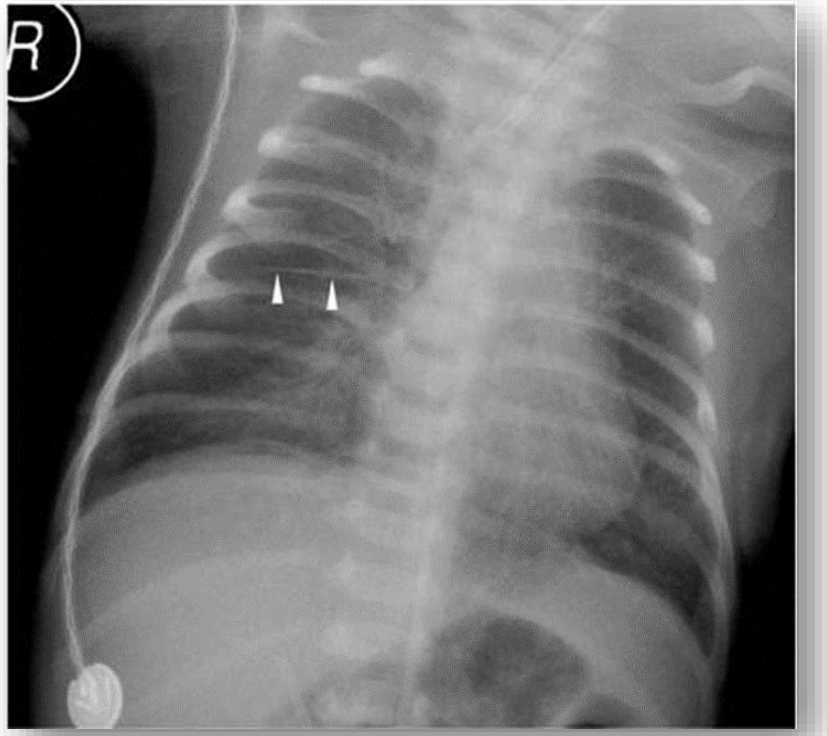
Transient Tachypnea of the newborn (TTN)



- **Term**: Most commonly full-term
- **Risk factors**: C-section
- **Onset of symptoms**: Immediately after birth
- **Clinical features**
  - Tachypnea
  - Increased breathing effort
  - Diffuse crackles, diminished, or normal breathing sounds on auscultation
- **Imaging**
  - Fluid in the lung fissures and increased lung volumes
- **Treatment**
  - Supportive care (supplemental oxygen, neutral thermal environment, adequate nutrition)

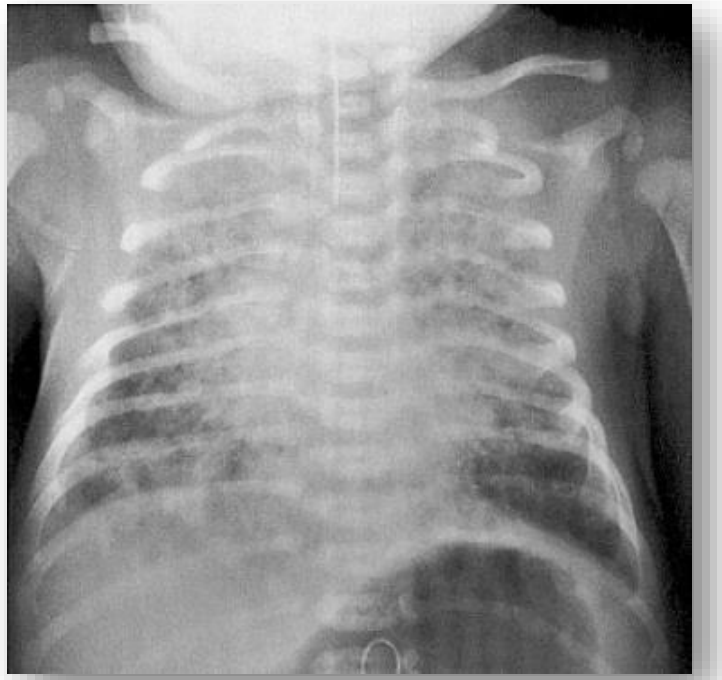
A full-term newborn baby born by c-section with this x-ray

- **What is your diagnosis ?**
  - Transient Tachypnea of the newborn (TTN)
- **Xray findings**: Fluid in the lung fissures and increased lung volumes
- **What is the treatment ?**
  - Supportive care (supplemental oxygen, neutral thermal environment, adequate nutrition)
- **Prognosis**: Resolves without complications in the majority of cases



Meconium Aspiration

- **Term**: Most commonly post-term
- **Risk factors**: Post-term delivery, Oligohydramnios
- **Onset of symptoms**: Immediately after birth
- **Clinical features**
  - Green amniotic fluid, Low APGAR score, Tachypnea, Increased breathing effort, Hypoxia, Lung rales and rhonchi
- **Imaging**
  - Increased lung volumes (increased anteroposterior diameter)
  - Asymmetric, patchy opacities
  - Flattening of the diaphragm
  - Pleural effusion



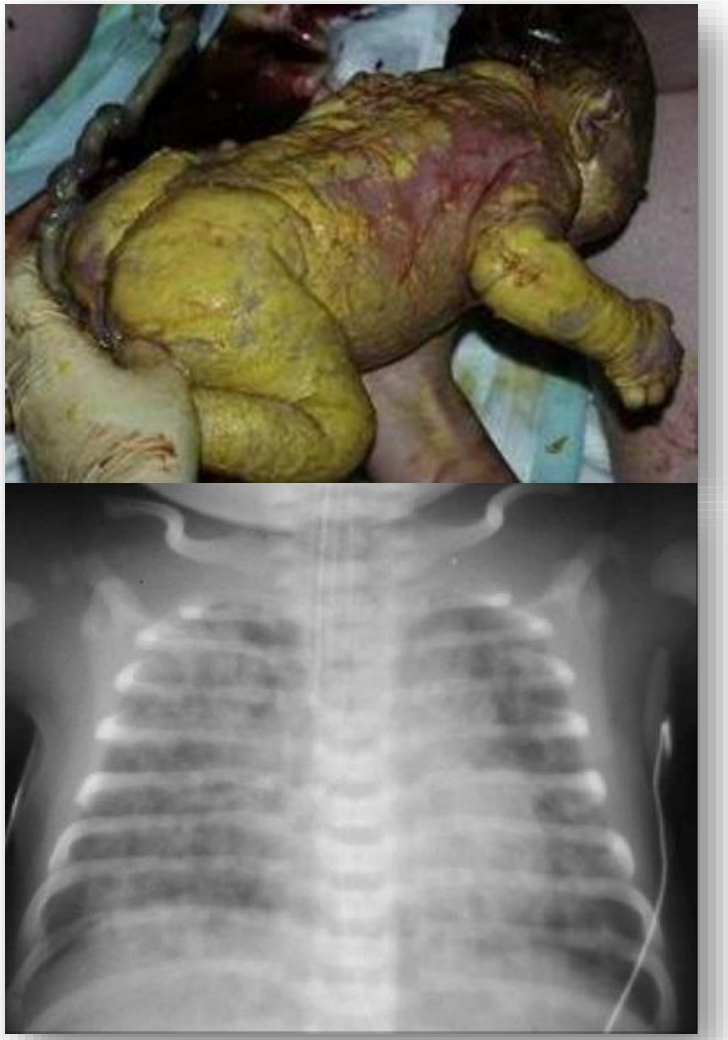
Meconium Aspiration

❖Management of meconium aspiration

- **Vigorous**: Defined as normal respiratory effort, normal muscle tone, and heart rate >100 bpm
  1. Do not electively intubate
  2. Clear secretions and meconium from the mouth and nose with a bulb syringe or a large bore suction catheter
- **Non- vigorous**: defined as depressed respiratory effort, poor muscle tone, and/or heart rate <100 bpm
  - Use direct laryngoscopy, intubate, and suction the trachea immediately after delivery.
  - Suction for no longer than 5 seconds.
  - Suction before his first breath

Meconium Aspiration

- **What is your diagnosis ?**
  - Meconium aspiration
- **Initial management if the baby is vigorous ?**
  1. Do not electively intubate
  2. Clear secretions and meconium from the mouth and nose with a bulb syringe or a large bore suction catheter
- **Mention 2 complications**
  - ARDS &
  - Persistent pulmonary HTN
  - Pneumothorax
  - Pneumomediastinum



Neonate with these findings and non-vigorous

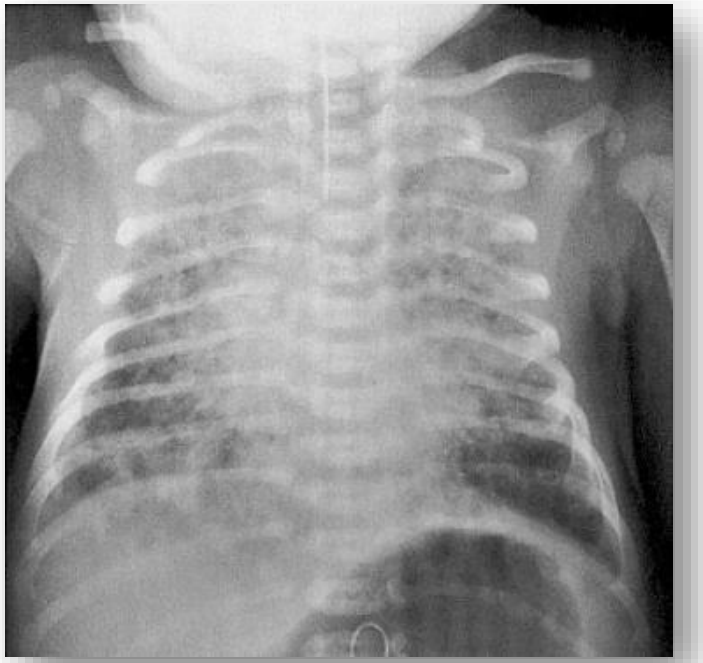
❖ **What is the first step in management ?**

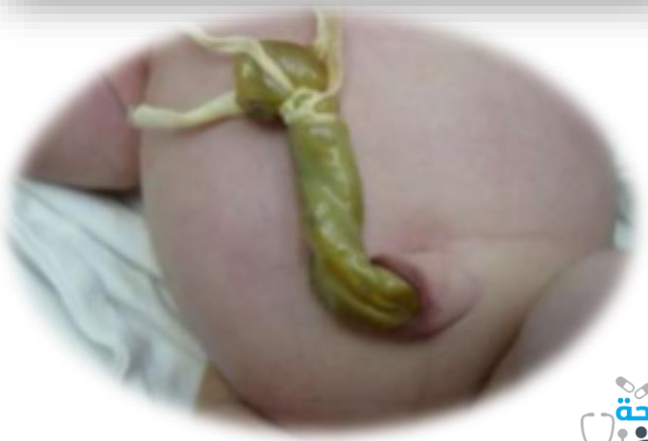
- Use direct laryngoscopy, intubate, and suction the trachea immediately after delivery.
- Suction for no longer than 5 seconds.
- Suction before his first breath

➢ If no meconium is retrieved, do not repeat intubation and suction.

➢ If meconium is retrieved and no bradycardia, HR <100 is present, > reintubate and suction.

➢ If the heart rate is low, administer positive pressure ventilation and consider suctioning again later.





Diaphragmatic hernia

- Newborn presented with respiratory distress & scaphoid abdomen. X-ray of patient is shown

❖ **Mention 2 prominent findings.**

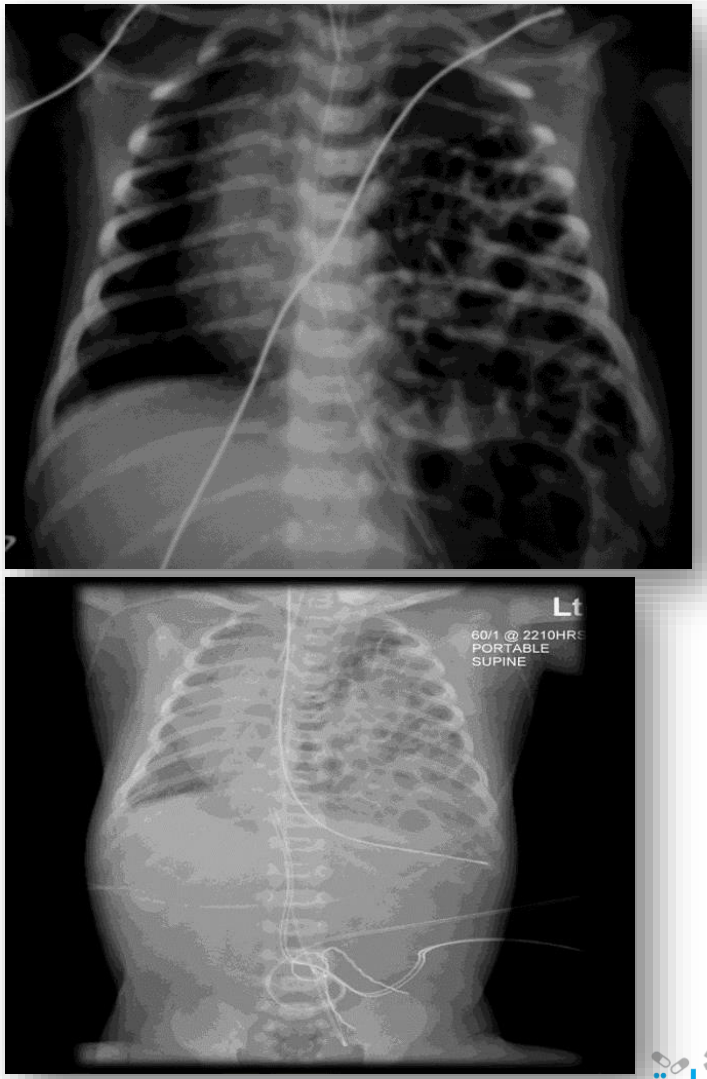
- Gas at Lt. chest side, Tracheal deviation to Rt.

❖ **What is the diagnosis ?**

- Diaphragmatic hernia

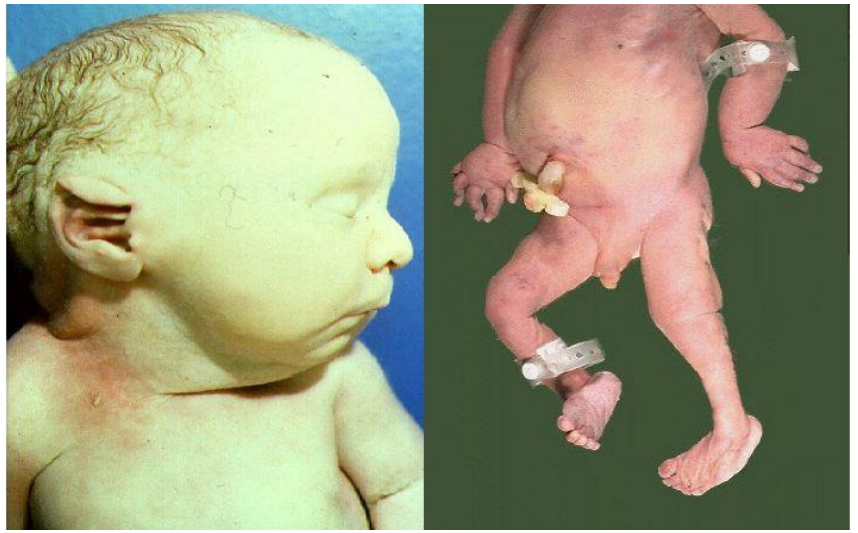
❖ **What is your next step for management ?**

1. Respiratory support with intubation and ventilation
2. A nasogastric tube should be passed for decompression

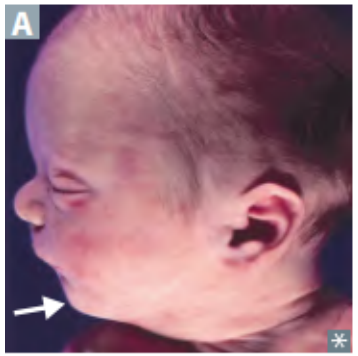


Pulmonary hypoplasia

- A pregnant complained of oligohydramnios and had this baby
- **What is the diagnosis ?**
  - Potter sequence



Potter sequence



Oligohydramnios → compression of developing fetus → limb deformities, facial anomalies (eg, low-set ears and retrognathia A, flattened nose), compression of chest and lack of amniotic fluid aspiration into fetal lungs → pulmonary hypoplasia (cause of death).

Caused by chronic placental insufficiency or reduced renal output, including ARPKD, obstructive uropathy (eg, posterior urethral valves), bilateral renal agenesis.

Babies who can't "Pee" in utero develop Potter sequence.

POTTER sequence associated with:

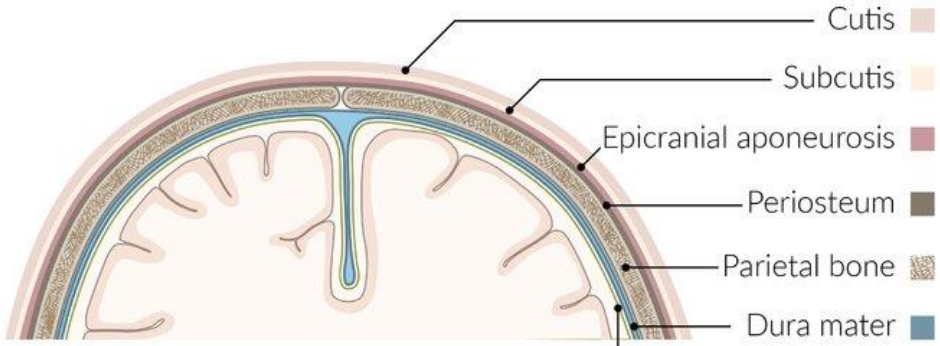
Pulmonary hypoplasia
Oligohydramnios (trigger)
Twisted face
Twisted skin
Extremity defects
Renal failure (in utero)

Birth injuries

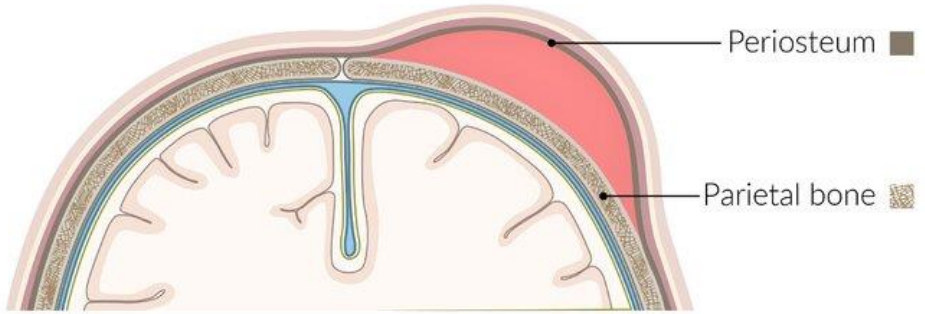
Medium-Yield

Neonatal soft tissue injuries

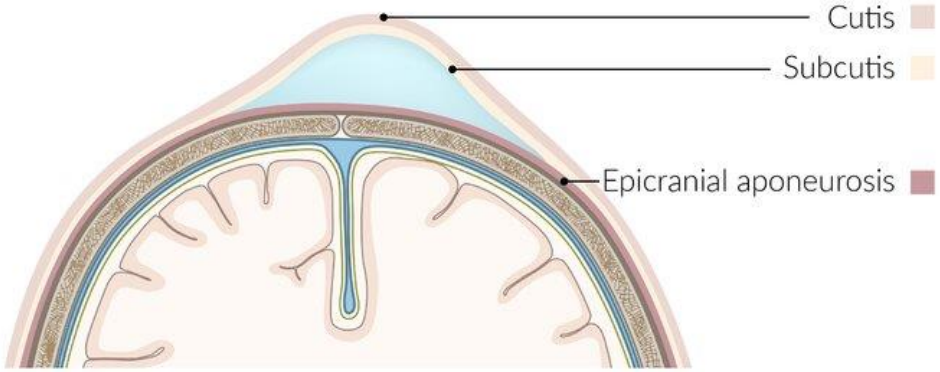
- **Head molding**
  - Transient deformation of the head into an elongated shape due to external compression of the fetal head as it passes through the birth canal during labor
  - Typically resolves within a few days after the birth
- **Caput succedaneum**: benign edema of the scalp tissue that extends across the cranial suture lines
  - Firm swelling; pits if gentle pressure is applied
  - No treatment required; resolves within hours or days
- **Cephalohematoma**: subperiosteal hematoma that is limited to cranial suture lines
  - Complications: calcification of the hematoma, secondary infection
  - No treatment required; resolves within several weeks or months
- **Subgaleal hemorrhage**
  - Rupture of the emissary veins and bleeding between the periosteum of the skull and the aponeurosis that may extend across the suture lines
  - Associated with a high risk of significant hemorrhage and hemorrhagic shock



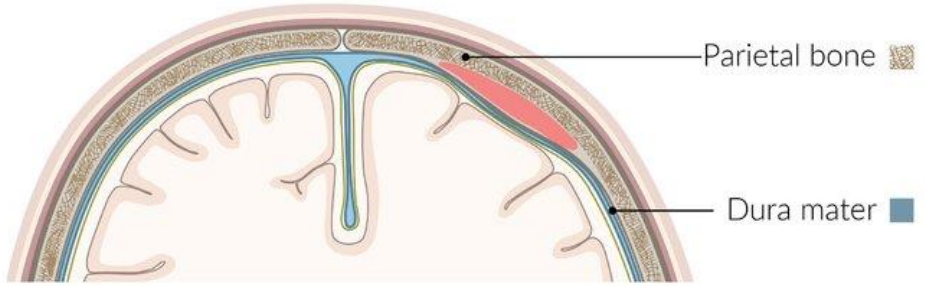
Normal



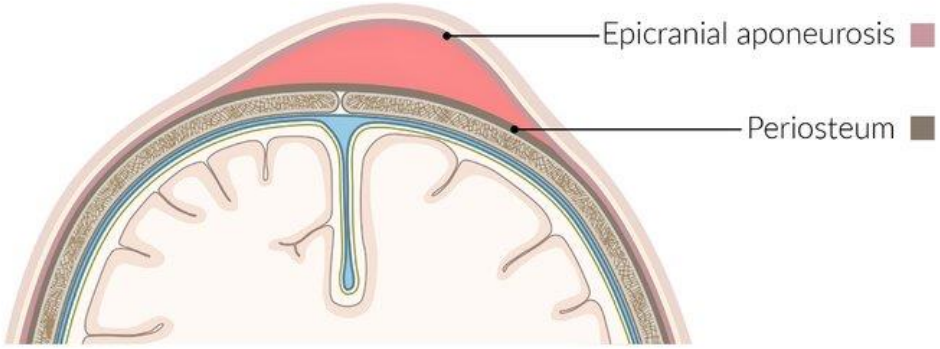
Cephalohematoma



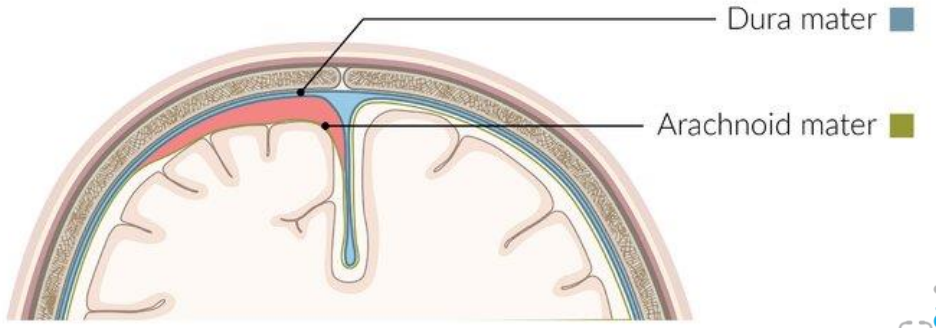
Caput succedaneum



Epidural hemorrhage



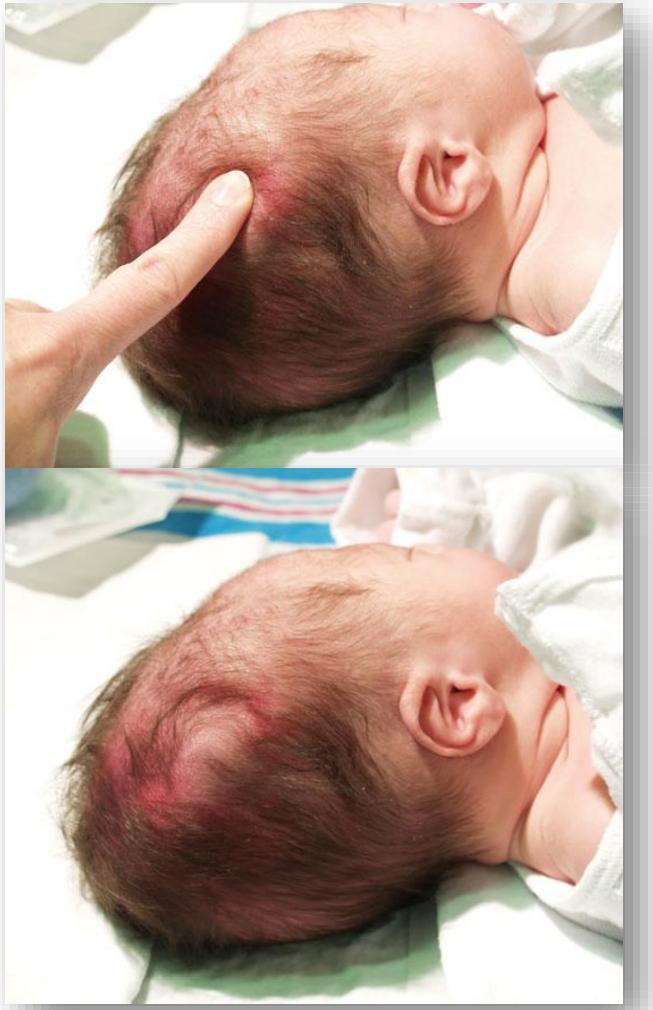
Subgaleal hematoma



Subdural hemorrhage

Caput succedaneum

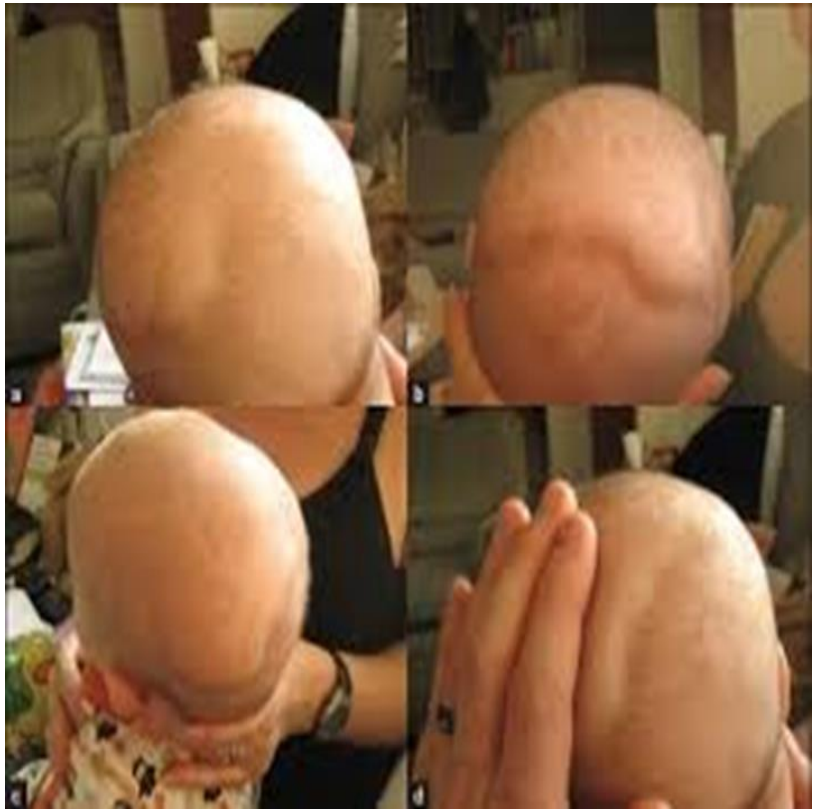
- **Name the finding you see in this newly born infant ?**
  - Pitting swelling
- **What is your diagnosis ?**
  - Caput succedaneum
- **Mention one complication that may occur in this patient**
  - Prolonged neonatal jaundice
- **Mention another DDx for this case.**
  - Cephalohematoma



CAPUT SUCCEDANEUM	CEPHAL HAEMATOMA
1. Present at birth on normal vaginal delivery.	1. Appears within a few days after birth on normal or forceps delivery.
2. May lie on sutures, not well defined.	2. Well defined by suture, gradually developing, hard edge.
3. Soft, pits on pressure.	3. soft, elastic but does not pits on pressure.
4. Skin ecchymotic.	4. No skin change.
5. Size largest at birth , gradually subsides within a day.	5. Become largest after birth and then disappears within 6-8 weeks to few months.
6. No underlying skull bone fracture.	6. May underlying skull bone fracture.
7. No treatment required.	7. No treatment required.

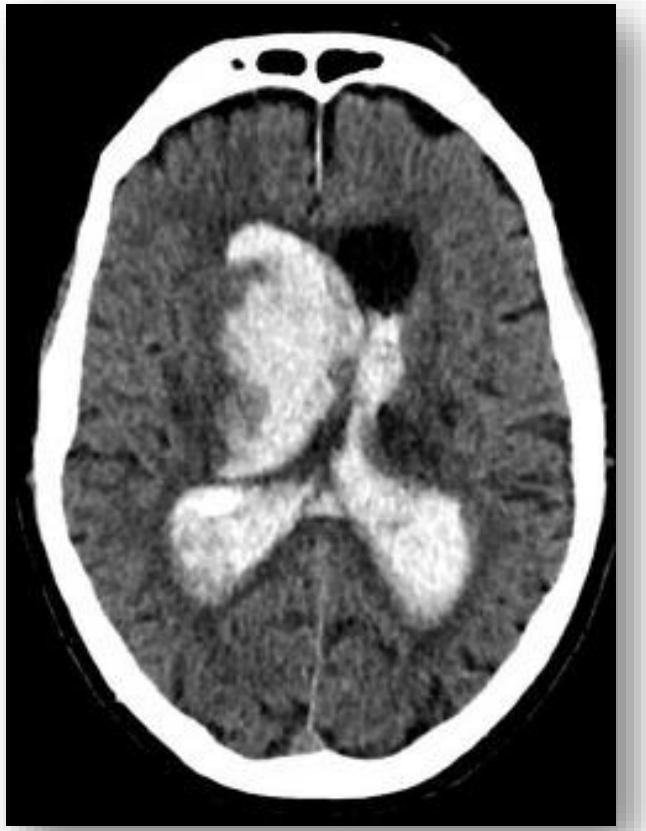
Subgaleal hematoma

- Long description of the hematoma and it’s extension
- **What is your diagnosis ?**
  - Subgaleal hematoma
- **Mention 2 lines of management**
  - Strict observation for complication
  - Appropriate resuscitation
  - Monitor bilirubin levels



Intraventricular hemorrhage

- **What findings are seen in this picture ?**
  - Intraventricular hemorrhage
- **Mention 2 complications of this conditions ?**
  - Periventricular leukomalacia
  - Developmental impairment
  - Double diplegic spastic CP
  - Seizures

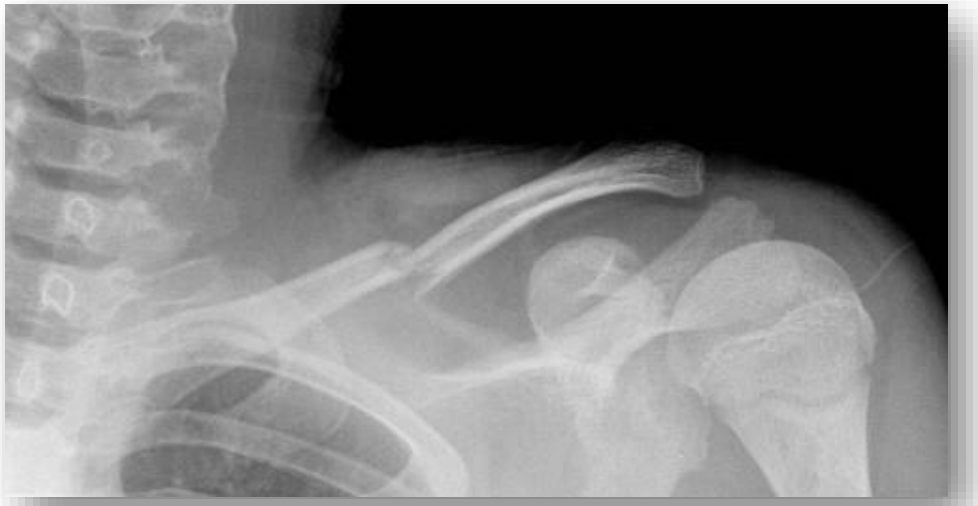


Birth-related clavicle fracture

❖ **What is your diagnosis ?**
- Left clavicle fracture

❖ **Write 2 clinical manifestation in this infant**
- Asymmetrical Moro reflex, brachial plexus injury

❖ **Mention 2 complications**
- Erb's palsy, Klumpke palsy



- Epidemiology: most common fracture during birth
- May be associated with macrosomia
- Usually, asymptomatic
- Diagnosed clinically: Bone irregularities, crepitus, and tenderness over the clavicle possible on palpation
- Usually self-resolves within 2–3 weeks without surgical intervention or long-term complications

Facial nerve palsy due to birth trauma

❖**What are the findings ?**

- 1) asymmetrical facies with crying.
  2) The mouth is drawn towards the normal side,
  3) wrinkles are deeper on the normal side

❖**What is your diagnosis ?**

- Left LMN lesion of facial nerve

➢**Epidemiology**: most common cranial nerve injury during birth

➢**Etiology**: Injury occurs during forceps-assisted delivery (most common)

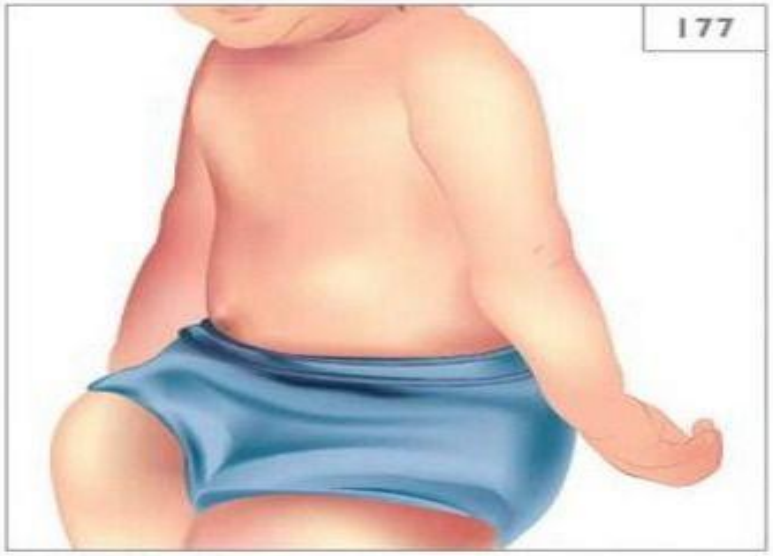
➢**Clinical features**: Peripheral facial nerve palsy : difficulty feeding, incomplete eye closure, absent nasolabial fold

➢**Treatment**: eye care with artificial tears and ointment

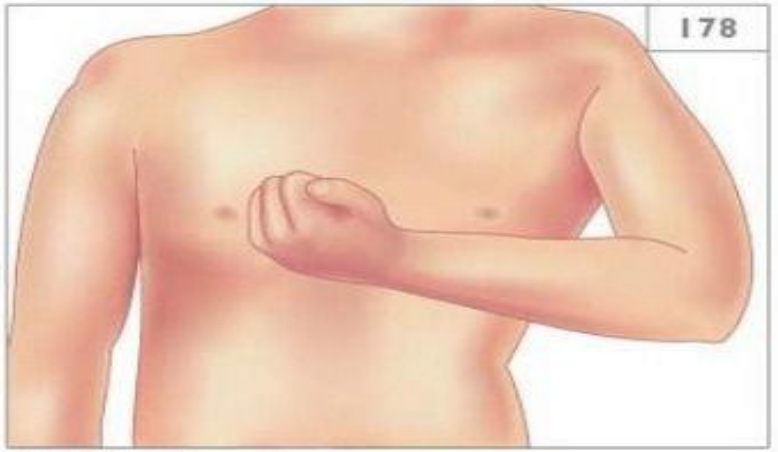
➢**Prognosis**: spontaneous recovery in 90% of cases within several weeks

Neonatal brachial plexus palsy

- Excessive lateral traction on the neck during delivery → injury to the upper trunk of the brachial plexus → Erb's palsy (C5,C6) (most common iatrogenic brachial plexus injury during delivery)
- Excessive traction on the arm during delivery → injury to the lower trunk of the brachial plexus → Klumpke palsy (C7, C8, T1)



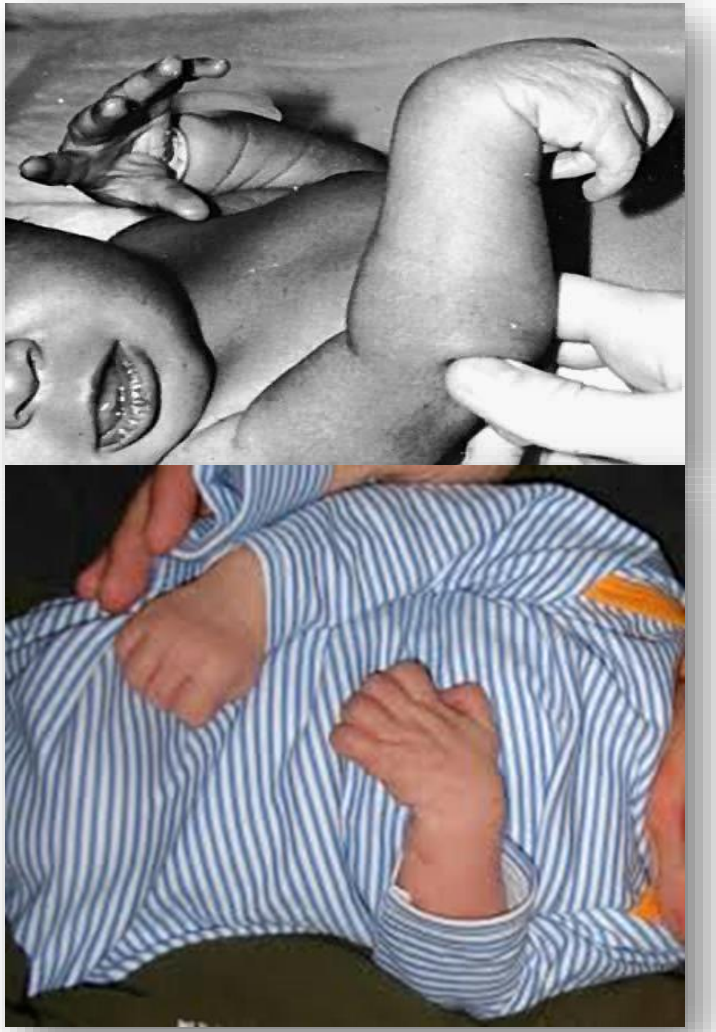
177 Erb's palsy.



178 Klumpke palsy.

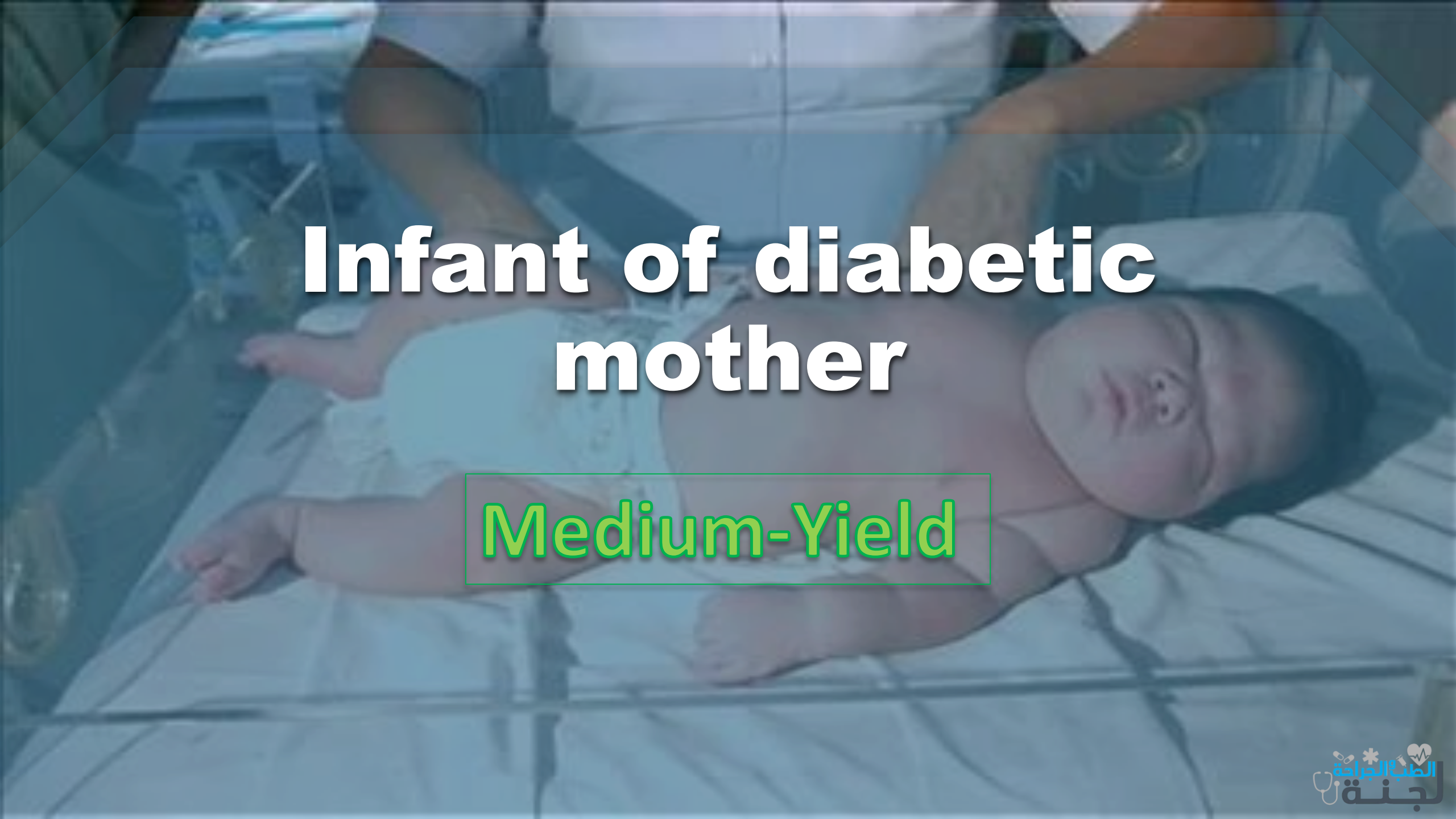
Neonatal brachial plexus palsy

- **What's the injured nerve ?**
  - Brachial plexus (C7, C8, T1)
- **What's the name of this deformity ?**
  - Klumpke palsy
- **What are the root that are affected ?**
  - C7, C8, T1
- **Risk factors for this condition ?**
  - Large baby
  - Breech delivery (bottom first)



Neonatal brachial plexus palsy

- **What do you see in the picture ?**
  - Asymmetrical Moro reflex
- **Mention 2 causes for this condition**
  - Erb's palsy
  - Humerus fracture
  - Clavicle fracture



Infant of diabetic mother

Medium-Yield

Complications in infants of diabetic mother

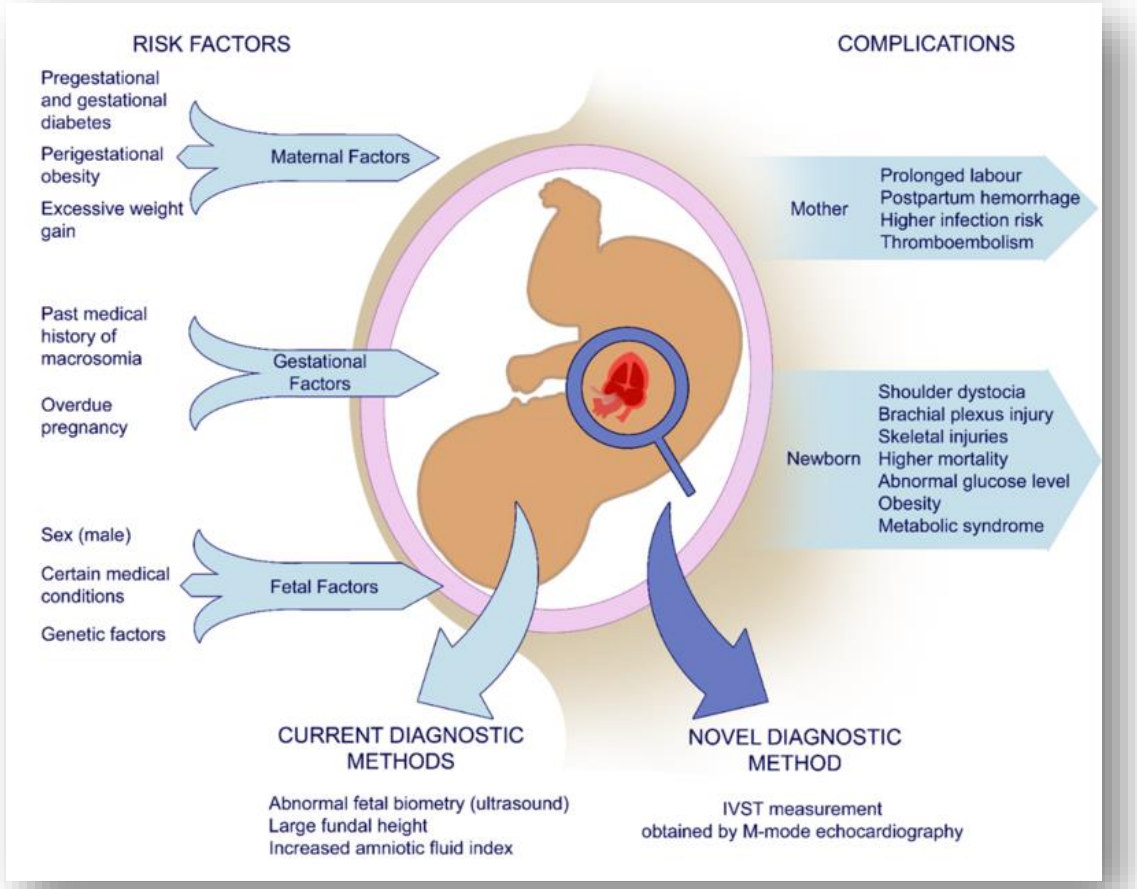
1. IUGR: Intrauterine growth restriction
   - Associated with pregestational DM
2. Fetal macrosomia (Large for Gestational age (LGA))
3. Hypoglycemia
4. Hematologic problems
   - Thrombocytopenia, Polycythemia, Hyperbilirubinemia
5. Hypocalcemia and hypomagnesemia
6. Pulmonary disease
   - Insulin blocks the development of enzymes necessary for the synthesis of lecithin
7. Cardiovascular anomalies
   - Transient hypertrophic cardiomyopathy > VSD > TGA > Single umbilical artery
8. Congenital malformations
   - Neural tube defects, Renal anomalies, GI defects (duodenal atresia, Anorectal atresia, small left colon syndrome)

Intrauterine growth restriction – Types

- **Asymmetrical IUGR**:
  - Predominantly occurs in the third trimester, affecting the growth rather than the formation of organs.
  - The brain develops normally while muscle mass, subcutaneous fat, and abdominal organs (liver) remain small.
- **Symmetrical IUGR:** global growth restriction
  - The entire body is proportionally small.
  - The circumference of the head is proportional to the rest of the fetal body.
  - ↑ Risk of neurologic sequelae; because growth restriction begins early in pregnancy, neural development, which is usually complete by the 18th week of gestation, may be affected.
- Pregestational DM is associated with symmetrical IUGR which result in a baby being Small for Gestational age (SGA)

Fetal macrosomia

- A significantly larger-than-average fetus, defined as birth weight > 90th percentile or > 4,000 g (8 lbs 13 oz)
- Associated with an increased risk of birth injuries
- Most commonly due to maternal diabetes
  - Maternal hyperglycemia → fetal hyperglycemia → stimulation of fetal pancreas → fetal hyperinsulinemia → ↑ hepatic glucose uptake and glycogen synthesis → ↑ fat and protein synthesis



Newborn baby, birth weight 4.5 kg

- **Mention 2 causes of this pathology**
  - Pregestational and gestational diabetes
  - Pregestational obesity
  - Excessive weight gain during pregnancy
  - History of previous macrosomia
- **Mention 2 complications of this pathology**
  - Shoulder dystocia
  - Brachial plexus injuries
  - Skeletal injuries
  - Higher mortality

Hypoglycemia, Polycythemia & Electrolytes abnormalities

❖Neonatal hypoglycemia

- Maternal hyperglycemia → chronic fetal hyperglycemia → ↑ metabolic effects and oxygen demand → fetal hypoxemia → ↑ erythropoietin concentrations → polycythemia

❖Neonatal polycythemia

- Associated with an increased risk of hyperviscosity syndrome, hyperbilirubinemia, and low iron stores
- ↓ Erythrocyte and iron storage and redistribution of fetal iron → iron deficiency

❖Electrolyte disturbances

- neonatal hypocalcemia (serum calcium < 7 mg/dL or serum ionized calcium < 4 mg/dL) and neonatal hypomagnesemia (serum magnesium < 1.5. mg/dL)
- Typically observed within 72 hours after birth
- ↑ Maternal urinary Mg excretion → hypomagnesemia in the mother and fetus → impaired PTH synthesis in the fetus → fetal hypocalcemia
- **Clinical features**: commonly asymptomatic but may manifest with lethargy, jitteriness, and/or seizures
- **Management**: IV calcium and/or magnesium in symptomatic infants

Baby was hypoglycemic his blood sugar 25mg/dL

- **Write other electrolytes abnormalities**
  - Hypomagnesemia, Hypocalcemia, Hyperphosphatemia
- **Write other causes of respiratory distress in this child, while HgA1c of his mother =9**
  - Congenital heart disease, TTN
- **Write 2 renal anomalies in this condition**
  - Hydronephrosis, renal agenesis, ureteral duplication
- **Write 2 neurological defects in this condition**
  - Neural tube defects (most common), Cauda equina regression syndrome (pathognomonic for IDM)
- **Write 2 cardiac anomalies in this condition**
  - Transient hypertrophic cardiomyopathy, TGA, truncus arteriosus, VSD, single umbilical artery

These infants are at an increased risk of neonatal respiratory distress and may present within the first few hours after birth with tachypnea, nasal flaring, intercostal retractions, and hypoxia and its due to inhibition of surfactant by the insulin, so the function and the concentration of the surfactant will be affected.

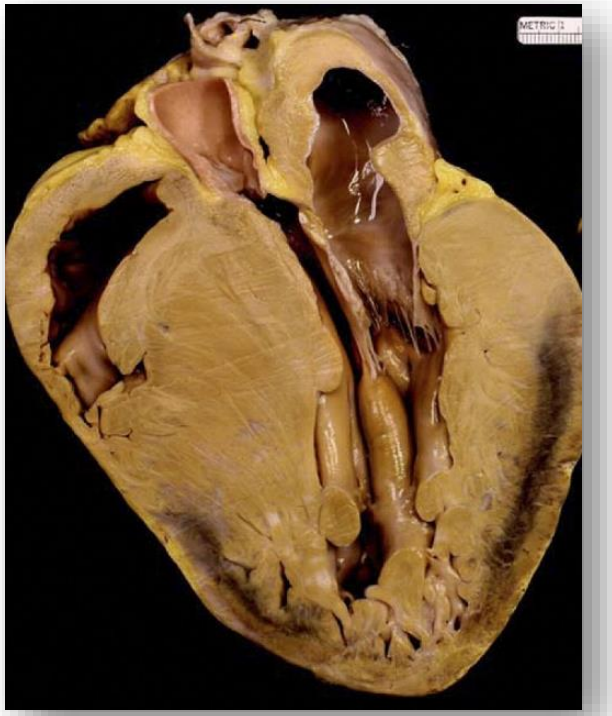
Macrosomia infant of diabetic mother

❖**What is the most common cause of the infant symptoms ?**

- Transient hypertrophic cardiomyopathy

❖**Write 2 GI malformation in this condition**

- Duodenal atresia, anorectal atresia, small left colon



Transient hypertrophic cardiomyopathy

➢**Clinical features**: often asymptomatic in infants but may manifest with symptoms of heart failure

➢**Diagnostics**: echocardiography

➢**Management**: supportive care

Small left colon syndrome

- 4.3 kg neonate presented with hypoglycemia and failure to pass meconium; barium enema was performed

❖ **What is the diagnosis ?**

- Small left colon syndrome

❖ **What is the underlying pathology ?**

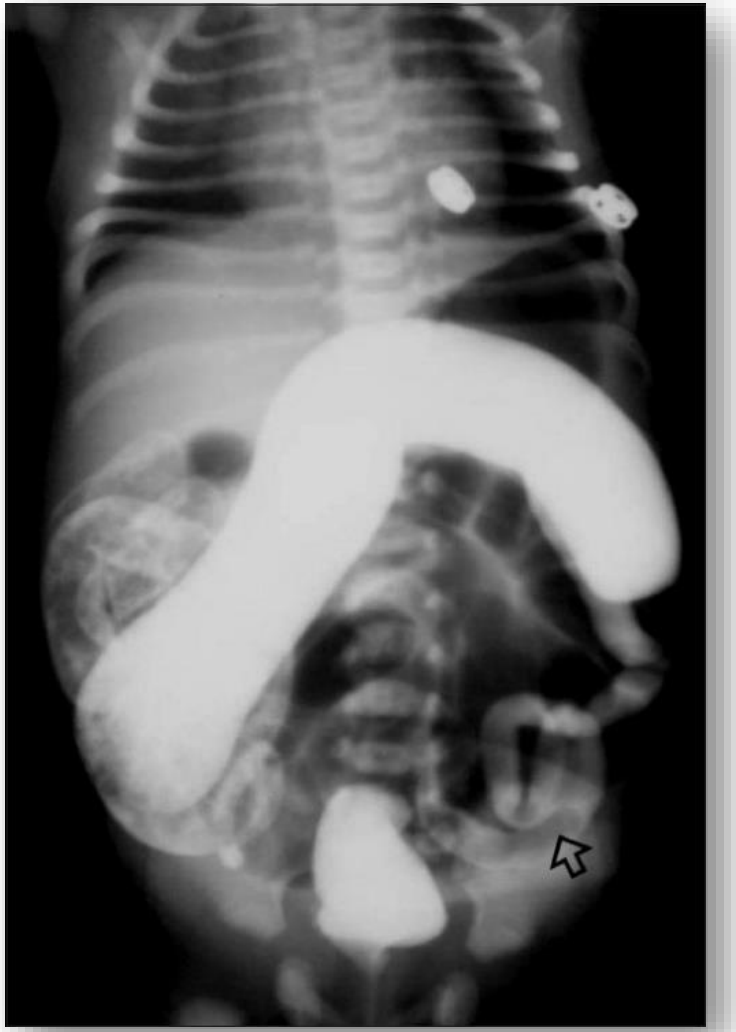
- Infant of diabetic mother

❖ **Mention another DDx for his condition**

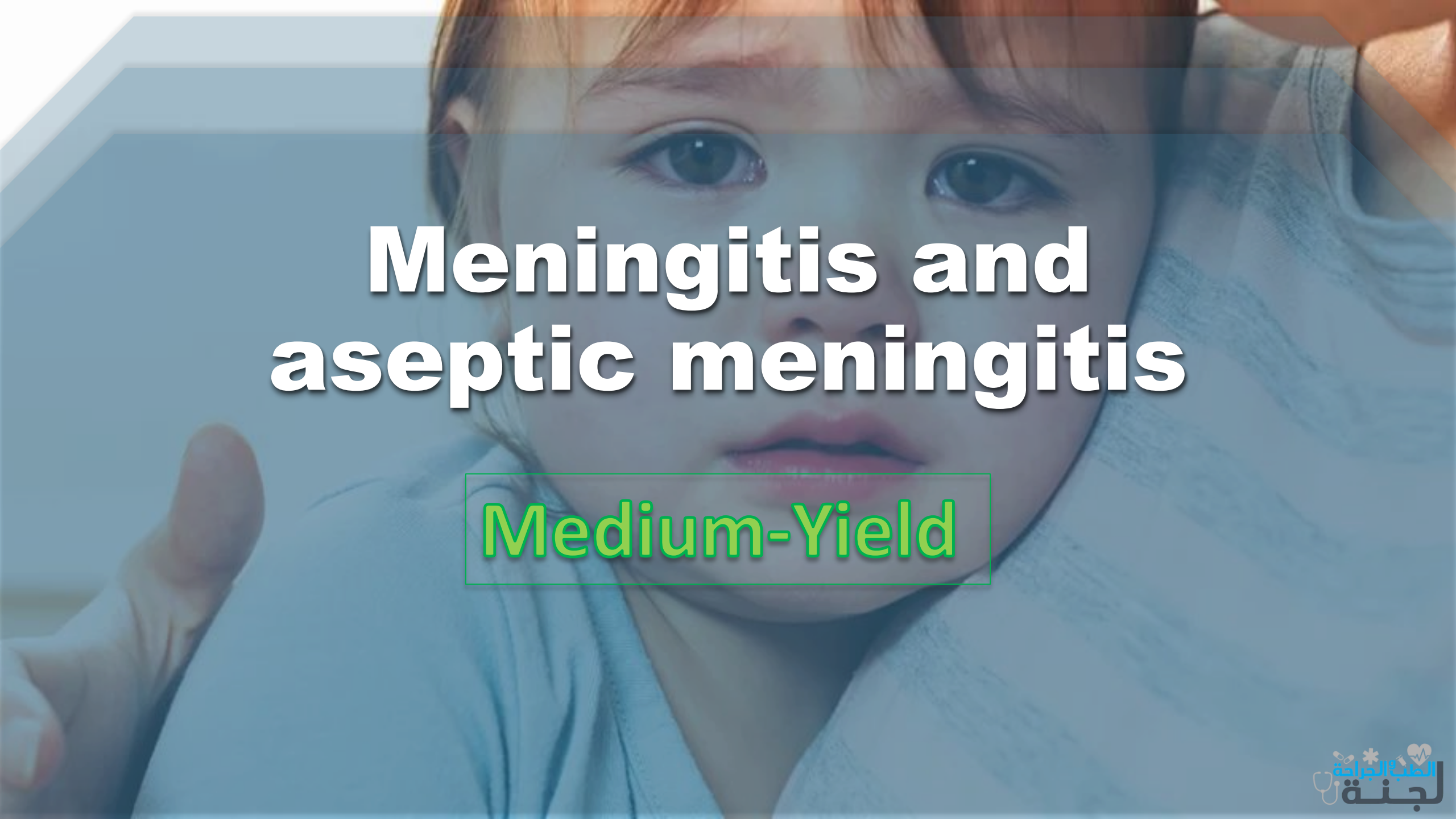
- Hirschsprung disease

❖ **What is the treatment ?**

- Conservative; if there is no perforation
- Contrast anemia is both diagnostic & curative



Neurology & Psychiatry



Meningitis and aseptic meningitis

Medium-Yield

شرح

Meningitis – Common pathogens

Patient population	Pathogen
By age	

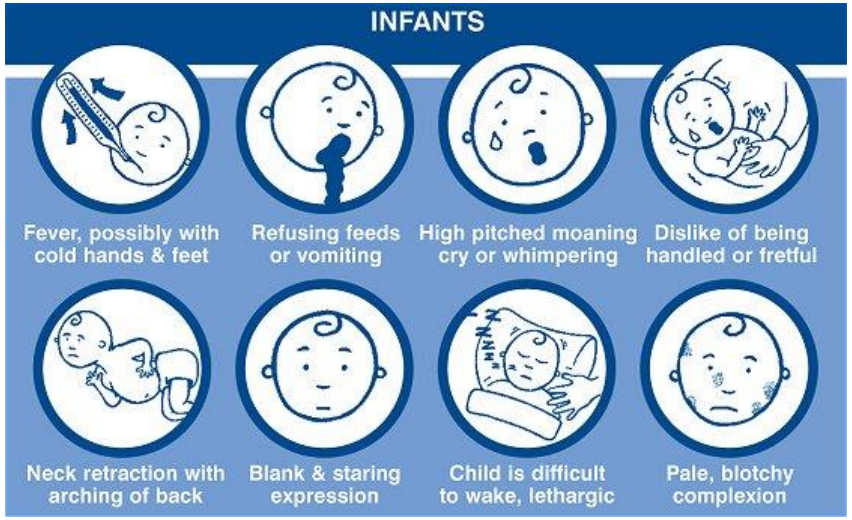
< 1 month [1]	• **Group B streptococcus** (esp. *Streptococcus agalactiae*)
	  ◦ The most common cause of bacterial meningitis and sepsis in neonates aged ≤ 72 hours [1]
	  ◦ The incidence of group B streptococcus infections in neonates is decreasing because of screening and prophylaxis during pregnancy. [6]
	• Gram-negative bacilli (e.g., *Escherichia coli*)
	• *Listeria monocytogenes*

1 month–2 years [2]	• ***Streptococcus pneumoniae*** • *Neisseria meningitidis* • Group B streptococcus (esp. *Streptococcus agalactiae*) • *Hemophilus influenzae* type b (if not immunized) [7]
2–50 years [2][8]	• ***Neisseria meningitidis*** (most common at the age of 11–17 years) • ***Streptococcus pneumoniae*** (most common in adults)

- Aseptic meningitis most commonly is a viral meningitis.
- Enteroviruses (especially coxsackieviruses and echoviruses): the most common cause of all types of meningitis in all patient groups

Name two differential diagnosis

- Meningitis
- Sepsis



- In neonates, meningitis often manifests with nonspecific symptoms and without the classic triad of meningitis.
  - The most important symptom is Poor feeding
- **Classic triad of meningitis**: fever, headache, and neck stiffness
  - Seen in children and adults

Diagnostics

- **What are the next steps in a child with suspected bacterial meningitis and NO alarm symptoms?**
  - Blood cultures → lumbar puncture → IV antibiotics (empirical treatment until the results of the culture then start guided treatment)
- **What are the next steps in a child with suspected bacterial meningitis and alarm symptoms?**
  - Blood culture + start empirical antibiotics/steroids → CT scan → LP to prevent herniation
- **What are important alarm symptoms in suspected meningitis?**
  - Signs of elevated ICP
  - Functional neurological disorder (e.g., weakness)
  - Altered mental status
  - Immunocompromised

Empirical treatment

- **Neonatal meningitis (<1-month)**
  - Ampicillin (for listeria) + Cefotaxime (for GBS & E.coli); Ceftriaxone is contraindicated in >1-month due to the risk of jaundice and kernicterus
- **Neonatal meningitis (>1-month – 90-days)**
  - Ampicillin (for listeria) + Cefotaxime OR Ceftriaxone (for GBS & E.coli)
  - Vancomycin is used in case of suspected highly resistant pneumococcus
- **Post-neonatal meningitis**
  - Dexamethasone (in case of S.pneumonae or meningococcemia)
  - 3rd generation cephalosporin (Cefotaxime OR Ceftriaxone) + Vancomycin
- **Meningitis in immunocompromised patients**
  - Ampicillin + Vancomycin + Cefepime or Meropenem

This procedure is used to rule out what ?

1. Meningitis or CSF infection
2. Central nervous system malignancies and paraneoplastic syndromes
3. Guillain-Barré syndrome
4. Multiple sclerosis
5. Neuroborreliosis
6. Subarachnoid hemorrhage (SAH)

CSF analysis – 5 tubes

1. **Cytology**: RBC: 0 /µL if present determine whither if it was traumatic or subarachnoid hemorrhage, WBC 0-5 /µL all lymphocytes
2. **CSF culture**
3. **Chemistry**: Protein: 20-40mg/dL, Glucose: 1/2 - 2/3 of blood glucose (mg/dL)
4. **Gram stain**
5. **Antigenicity**

	WBC /µL	Protein (mg/dL)	Glucose (mg/dL)
Normal	0-5 all lymphocytes	20-40	1/2 - 2/3 of blood glucose
Bacterial meningitis	100-10.000 Neutrophils	High (>100)	Decreased
Viral meningitis	Increased all lymphocytes	Mild elevation	Normal
TB / Fungal meningitis	Increased all lymphocytes	Very high	Very low
Demyelinating disease	Normal	High	Normal

CSF result for 5-day-old neonate who complained of vomiting

❖**What's your interpretation?**

- Leukocytosis, High neutrophiles, mild low protein & low glucose concentration

❖**What is the Diagnosis ?**

- Acute bacterial meningitis.

❖**Mention the most specific treatment.**

- Ampicillin + cefotaxime

- WBC: 155/µL
- Neutrophils: 70%
- RBC: 0/ µL
- Lymphocytes: 30%
- Serum Glucose: 5 mmol/L
- CSF glucose: 2 mmol/L
- Protein: 80 mg/dL

	WBC /µL	RBC /µL	Protein (mg/dL)	Glucose (mg/dL)
Neonates normal	Up to 25-30 all lymphocytes	Up to 100	90-190	1/2 - 2/3 of blood glucose

15 months old patient with history of vomiting and fever

❖**What is your DDx ?**
- Bacterial meningitis

❖**What are the most common 3 organism ?**
- S.pneumonae
- N.meningitidis
- H.infleunzae

❖**If the culture show gram + diplococci, what is the organism ?**
- S.pneumonae
- **Specific treatment**: Dexamethasone + Vancomycin

- Blood
  - WBC = 22, Platelets = 298, RBC = 4.5, Na = 136, K = 3.6, glucose = 95
- Urine:
  - RBC = 2-4, protein = +1, PH = 5.5
- CSF:
  - Protein = 110
  - Glucose = 2
  - WBC = 1000 (90 % neutrophile)

CSF result for 5 days neonate

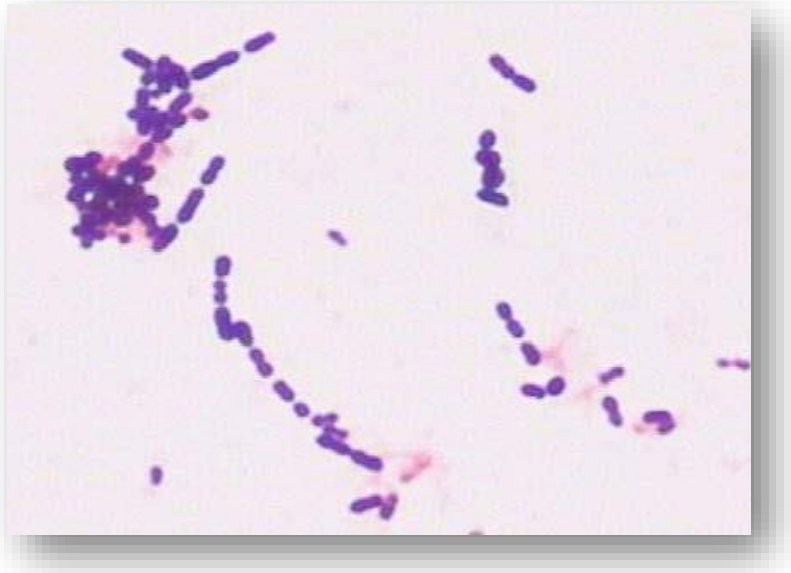
- **What is your interpretation ?**
  - Normal CSF “for neonate”.
- **Give 2 other CSF tests you will order**
  - Gram stain.
  - CSF Culture.
  - PCR.
  - Latex agglutination.

- WBCs = 22
- RBC = 0
- Proteins = 50
- Sugar = 3
- Blood sugar = 5

	WBC /µL	RBC /µL	Protein (mg/dL)	Glucose (mg/dL)
Neonates normal	Up to 25-30 all lymphocytes	Up to 100	90-190	1/2 - 2/3 of blood glucose

Gram stain for CSF for 5 y/o Pt with meningitis

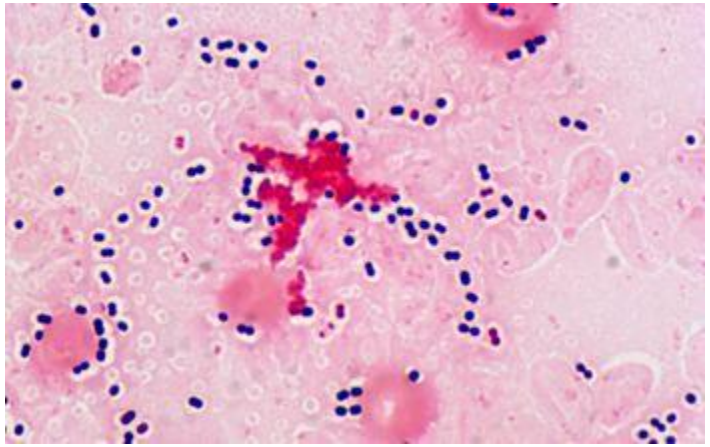
- **Identify the microorganism?**
  - Strep. Pneumonia
- **What is the treatment for it ?**
  - Corticosteroids + Vancomycin

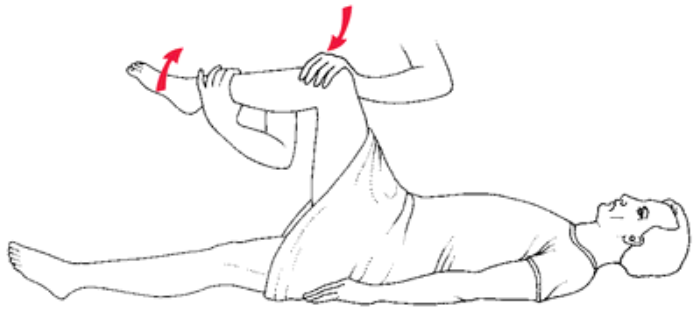


- Corticosteroids is given to prevent the neurological deficits associated with pneumococcal meningitis (e.g. deafness, focal deficits); should be discontinued if pneumococcal meningitis is ruled out

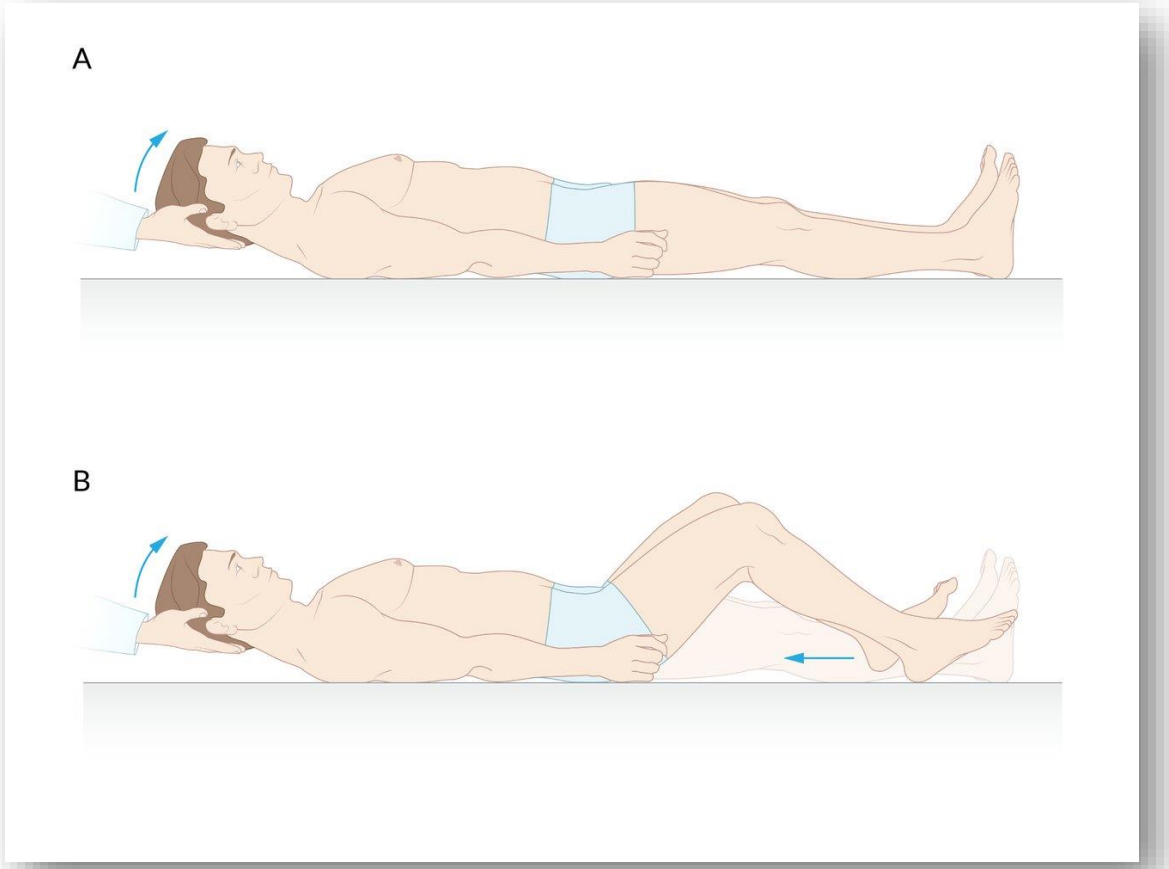
Presentation of meningitis of 6 months

- **What test is done here?**
  - CSF gram stain
- **What is the name of this sign ?**
  - Kernig’s sign
- **What is your management?**
  - Corticosteroids + Vancomycin (S.pneumonae)

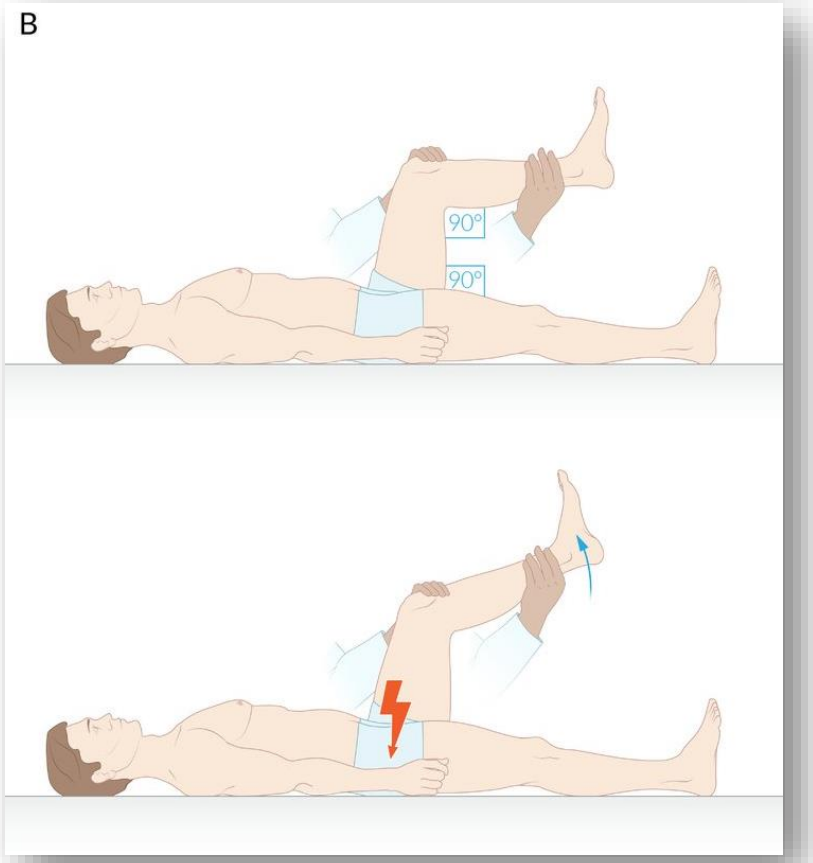




Signs of meningeal irritation



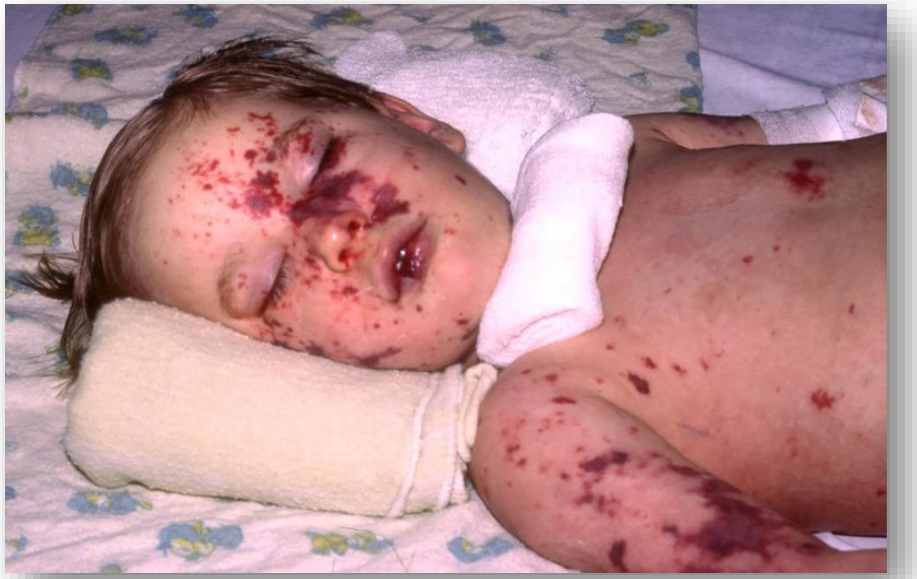
Brudzinski sign

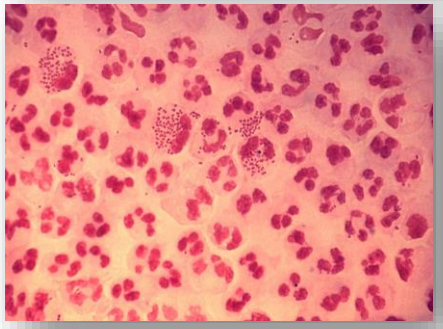


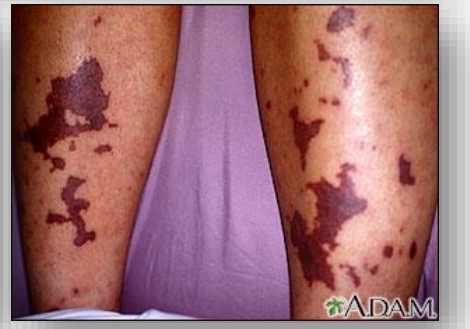
Kernig sign

Meningococcemia

- **What is your diagnosis?**
  - Meningococcemia
- **What's the causative organism ?**
  - Neisseria meningitidis
- **What dose it stain ?**
  - Gram negative
- **What are the lines of treatment ?**
  - 3rd generation cephalosporin
  - Steroids + dopamine + IV fluids

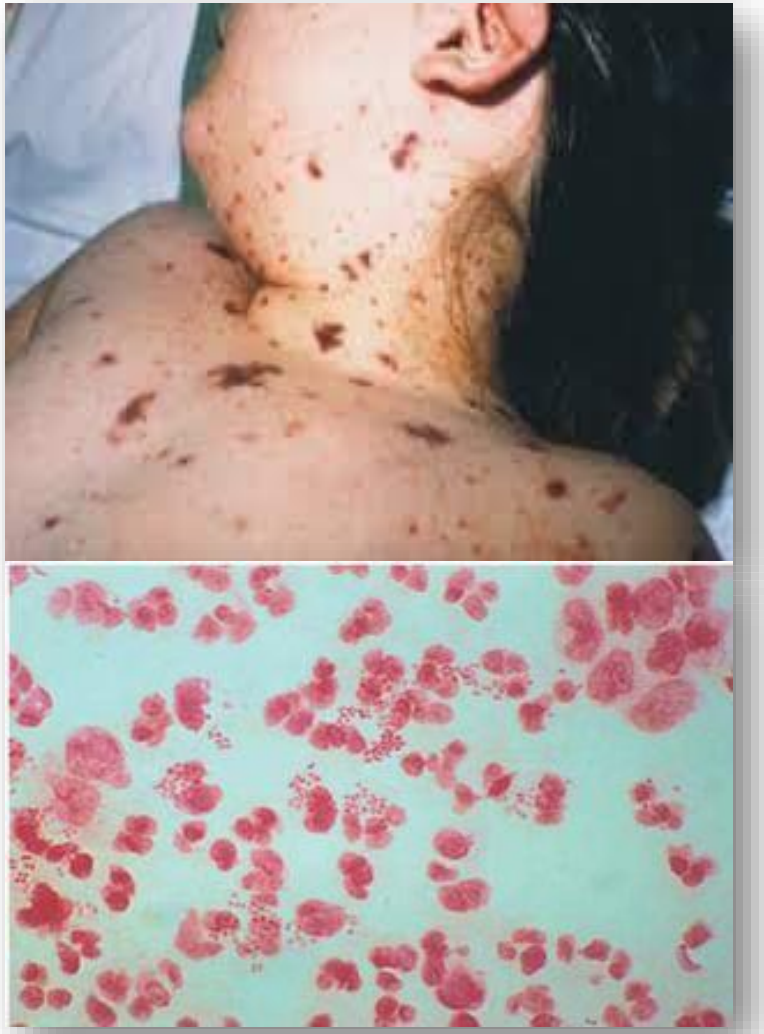






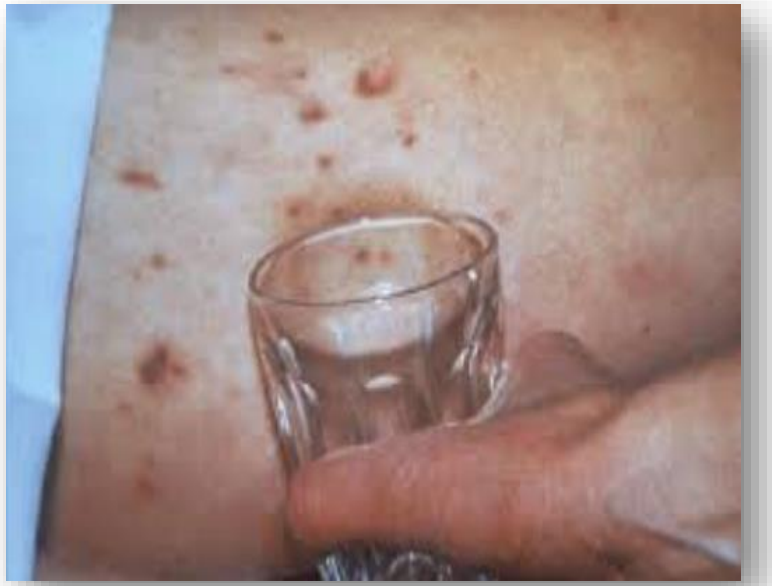
Presentation of meningitis of 1 year old

- **What is your diagnosis?**
  - Meningococcemia
- **What's the causative organism ?**
  - Neisseria meningitidis
- **What test is done here?**
  - CSF gram stain
- **What are the lines of treatment ?**
  - 3rd generation cephalosporin
  - Steroids + dopamine + IV fluids



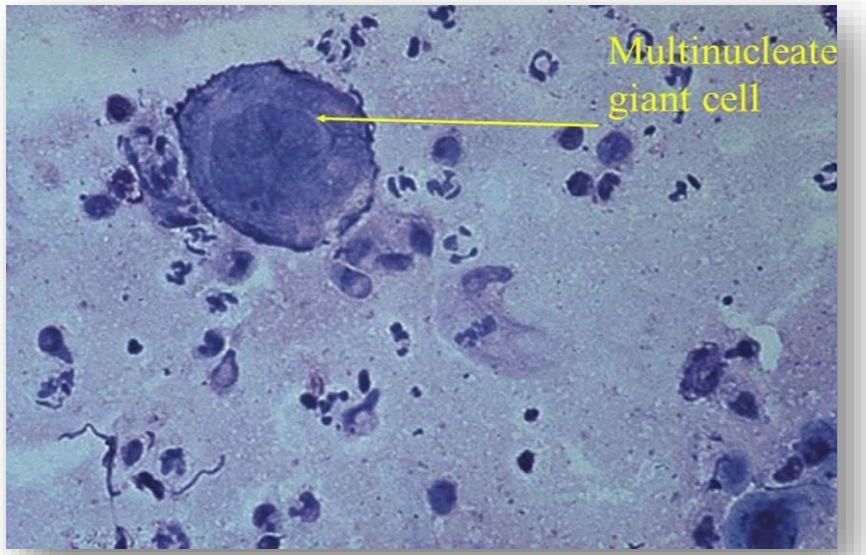
Skin glass test

- **Describe what do you see ?**
  - Skin glass test
- **What is it used for ?**
  - Differentiate between blanching / non-blanching purpura
- **Non-blanching purpura ddx:**
  - Meningococcemia & Thrombocytopenia



Case of HSV lesions

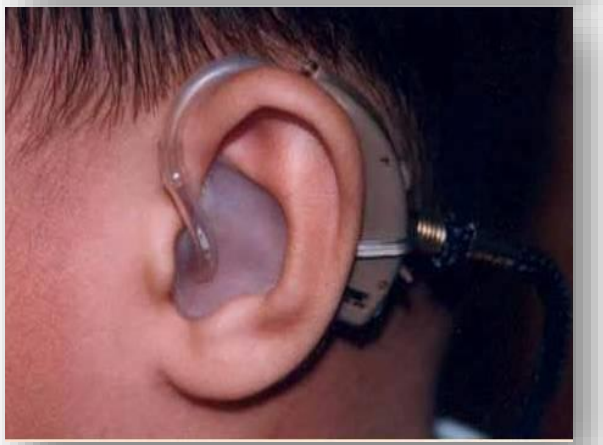
- **Name of the test and name of the finding**
  - Tzanck smear
- **What is your management and the duration of treatment ?**
  - Depend on the type of HSV in the scenario
- **What is the most affected lobe ?**
  - Temporal lobe
- **What is the treatment ?**
  - IV Acyclovir



Hearing difficulties

- **What is the aspect of developmental assessment is affected by this patient ?**
  - Hearing and speech and language
- **What is the infection at infancy cause this problem ?**
  - Meningitis (especially pneumococcal meningitis)

- **What's the abnormality in this child ?**
  - Hearing difficulties
- **What is other abnormality you think that you will find in this baby ?**
  - Cerebral palsy with speech difficulties
  - Epilepsy



Seizures

Low-Yield

Seizures

- **Absence**
  - idiopathic, familial
  - **Treatment**: ethosuximide (1st line) or valproic acid
- **West Syndrome**
  - Triad of infantile spasm, arrest psychomotor development, hypsarrhythmia
  - **Treatment**: ACTH, Prednisolone, or Vigabatrin
- **Juvenile Myoclonic Epilepsy**
  - idiopathic, familial
  - **Treatment**: Valproic acid (1st line)
- **Rolandic Epilepsy**
  - idiopathic, familial (age 5-12y)
  - **Treatment**: No treatment or carbamazepine (بتروح لحالها)

إضافي

Seizures

❖Febrile seizure

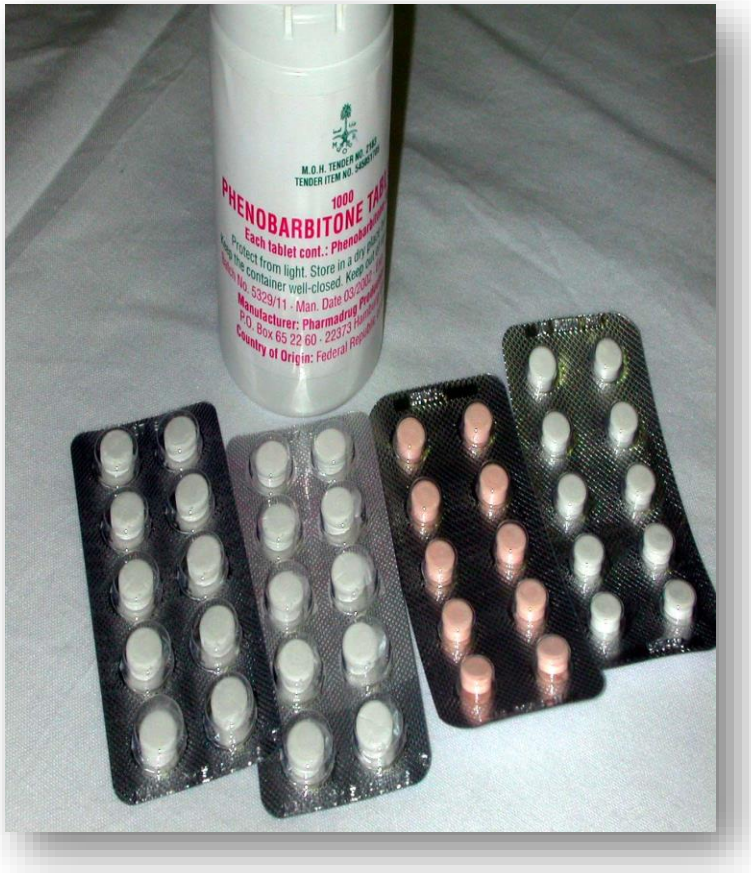
- Criteria: Seizure + fever, Age between 6m-5y, No evidence of CNS infection
- Complex seizure if duration >15m, multiple within 24h, or focal
- Common cause is roseola infection

❖Status epilepticus (seizures >5m) management

1. ABCDE
2. IV access
3. Early status 5-20m: Lorazepam or Diazepam
4. Established status 20-40m: Phenytoin → Phenobarbitone
5. Refractory status 40-60m: Midazolam; DONT use propofol in pediatrics
6. Super-Refractory status: Thiopental

Phenobarbitone (Luminal)

- **Uses (Broad spectrum)**
  - Neonatal seizures
  - Status epilepticus (Refractory)
- **S/Es**
  - Children: Hyperactivity, cognitive impairment
  - Adult: Sedation
  - Induction of cytochrome P-450
  - Cardiorespiratory depression



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Phenytoin (Epanutin)

- **Uses (Broad spectrum)**
  - Status epilepticus (Refractory)
- **S/Es** سنوات (1)
  - May reach toxic levels
  - Hirsutism
  - Coarse features
  - Hypertrophic gums
- **Notes**
  - Follows Zero-order pharmacokinetics
- Mnemonic: PPHENYTOIN
  - Cytochrome P-450 induction, Pseudo-lymphoma, Hirsutism, Enlarged gums, Nystagmus, Yellow-brown skin, Teratogenicity (fetal hydantoin syndrome), Osteopenia, Inhibited folate absorption, Neuropathy
- Rare: SJS, DRESS syndrome, drug-induced lupus.
- Toxicity leads to diplopia, ataxia, sedation

Ethosuximide (Zarontin)

- **Uses**
  - Absence epilepsy (1st line)
- **S/Es**
  - Agranulocytosis
  - Skin rash
- **Mnemonic**: EFGHIJ
  - Ethosuximide causes Fatigue, GI distress, Headache, Itching (and urticaria), SJS

Carbamazepine (Tegretol)

❖Uses (Narrow spectrum)

- Partial seizure
- Trigeminal neuralgia (1st line)
- Rolandic epilepsy
- Also used in complex seizures, and secondary generalization

❖S/Es

- Skin rash
- Blood dyscrasias (agranulocytosis, aplastic anemia)
- Hyponatremia (SIADH)

Valproic acid (Depakine)

❖Uses (Broad spectrum)

- Generalized seizure and Partial seizure
- Juvenile myoclonic epilepsy
- Absence seizure
- Mood stabilizer
- Migraine prophylaxis

❖S/Es

- Hepatotoxic
- Transient hair loss
- Weight gain

❖Notes

- A component in all anti-epileptic regimens
- When given with Lamotrigine, the dose should be lowered to ($^{1}/_{10}^{th}$) of the usual dose, since Valproic acid increases the half life of Lamotrigine

❖Mnemonic: VALPPROaTTE:

- Vomiting
- Alopecia
- Liver damage (hepatotoxic)
- Pancreatitis
- P-450 inhibition
- Rash
- Obesity (weight gain)
- Tremor
- Teratogenesis (neural tube defects)
- Epigastric pain (GI distress)

Vigabatrin (Sabril)

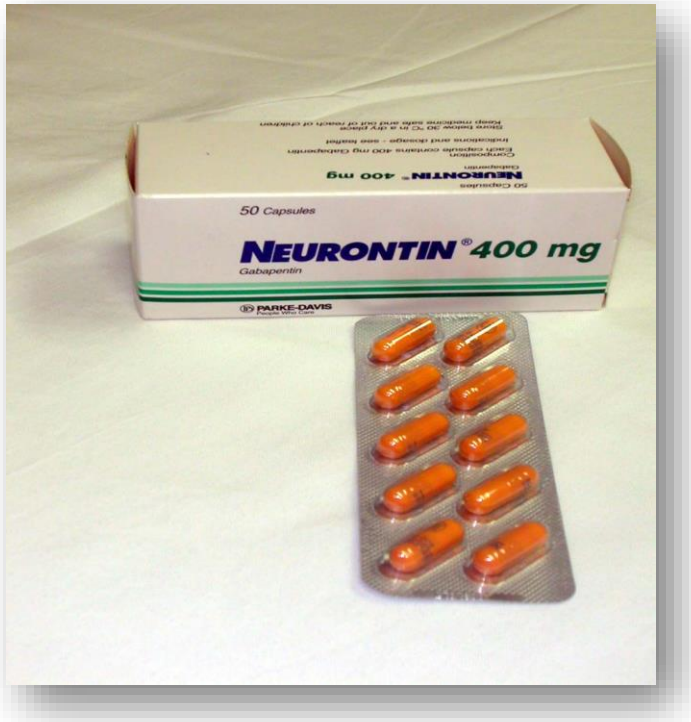
- **Uses (Narrow spectrum)**
  - West syndrome
  - Partial seizure
- **S/Es**
  - Visual field defects

إضافي

Gabapentin (Neurontin)

❖Uses (Broad spectrum)

- Partial seizure
- Neuropathic pain
  - Peripheral neuropathy
  - Postherpetic neuralgia



❖S/Es

- Sedation
- Ataxia

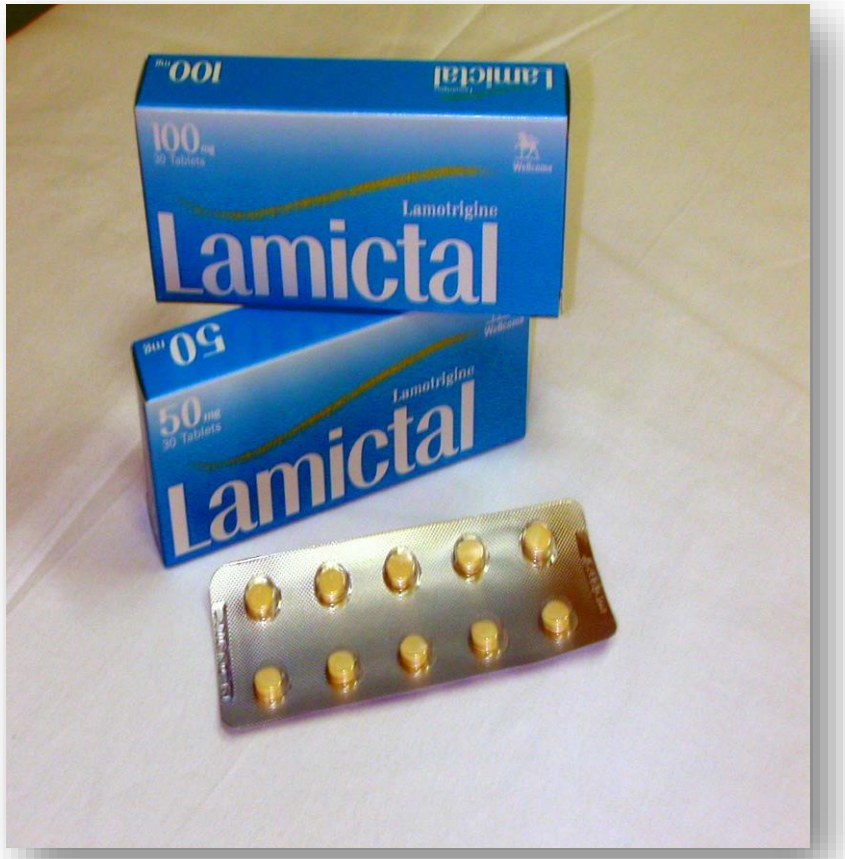
❖Notes:

- Has no interactions with other drugs
- Safe to be used in organ failure(Liver or renal failure)

إضافي

Lamotrigine (Lamictal)

- **Uses (Broad spectrum)**
  - Generalized seizure
  - Partial seizure
  - Absence seizure
- **S/Es**
  - Measles like rash
  - Stevens Johnson syndrome
  - Hemophagocytic lymphohistiocytosis (black box warning)



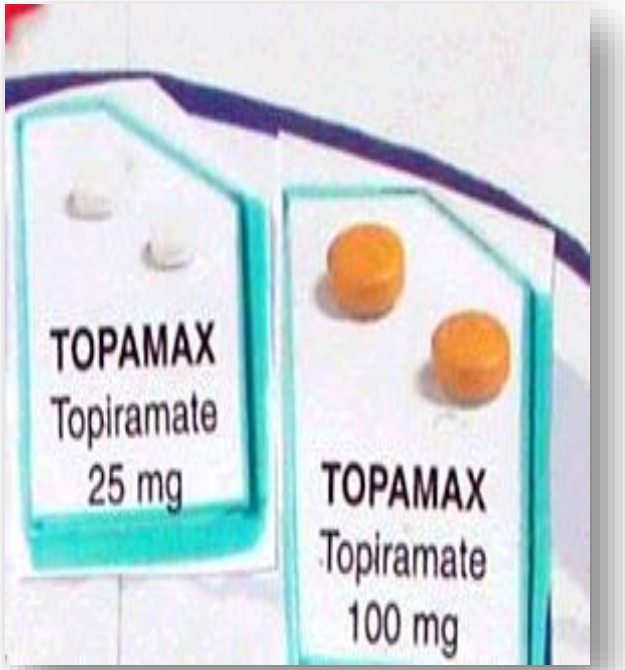
Topiramate (Topamax)

❖Uses (Broad spectrum)

- Main use: intractable partial complex seizures
- Migraine prophylaxis
- Other
  - Generalized seizure
  - Partial seizure

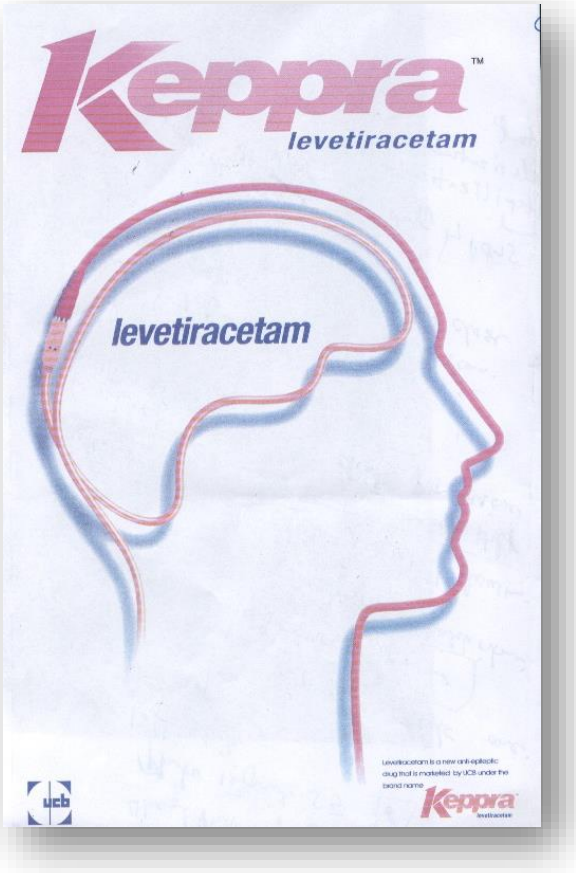
❖S/Es

- Renal stones
- Decreases attention and ability to concentrate
- Weight loss
- Acute angle closure glaucoma



Levetiracetam (Keppra)

- **Uses (Broad spectrum)**
  - Partial and tonic-clonic
- **S/Es**
  - Neuropsychiatric symptoms (eg, personality change)
  - Fatigue
  - Drowsiness
  - Headache
- **Notes**
  - Used as a primary drug.



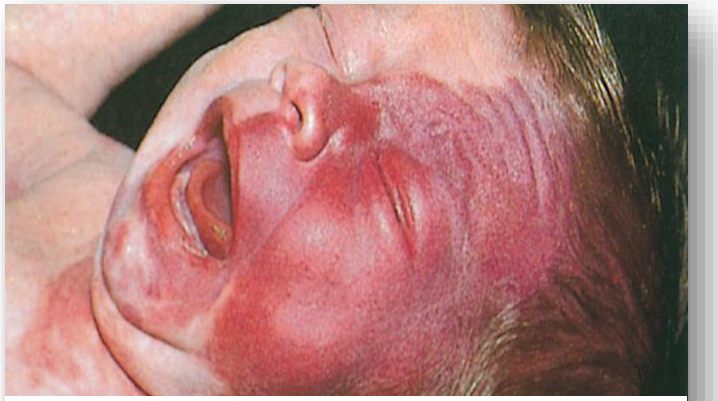
Neurocutaneous disorders

Low-Yield

المهم فقط الي بالأحمر الباقي زيادة

Sturge-Weber syndrome

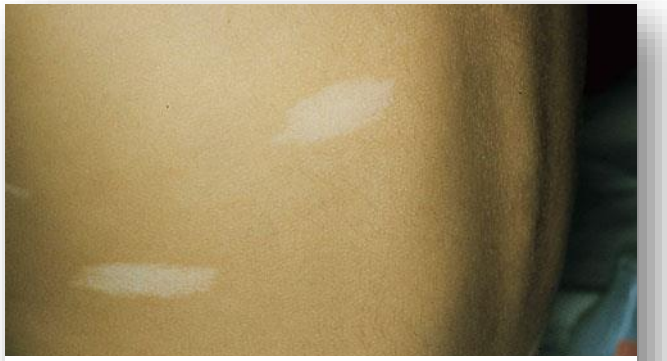
- The characteristics of Sturge-Weber syndrome may be remembered with the mnemonic "**STURGE**":
  - **S**: Sporadic, port-wine Stain
  - **T**: Tram track calcifications (opposing gyri)
  - **U**: Unilateral
  - **R**: Retardation (intellectual disability)
  - **G**: Glaucoma, GNAQ gene
  - **E**: Epilepsy



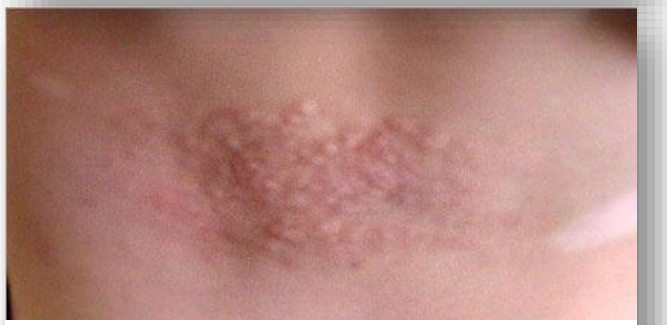
Port-wine stain

Tuberous Sclerosis

- The characteristics of tuberous sclerosis may be remembered with the mnemonic "**HAMARTOMAS**":
  - **H**: Hamartomas in CNS and skin
  - **A**: Angiofibromas (adenoma sebaceum)
  - **M**: Mitral regurgitation
  - **A**: Ash-leaf spots (hypopigmented macule)
  - **R**: cardiac Rhabdomyoma
  - **T**: Tuberous sclerosis
  - **O**: autosomal dOminant
  - **M**: Mental retardation (intellectual disability)
  - **A**: renal Angiomyolipoma
  - **S**: Seizures, Shagreen patches



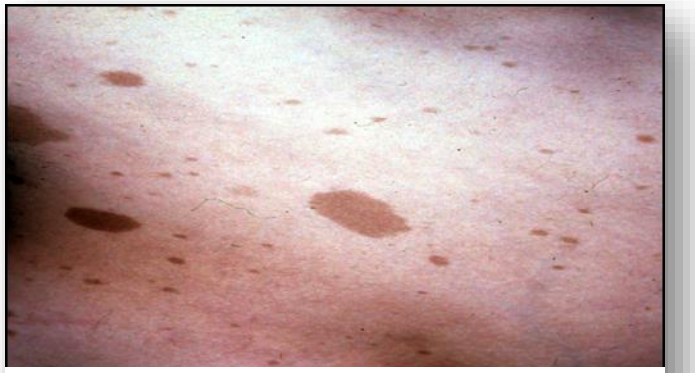
Ash-leaf spots



Shagreen patch

Neurofibromatosis type 1

- The characteristics of neurofibromatosis may be remembered with the mnemonic "**CICLOPSS**":
  - **C**: Café-au-lait spots
  - **I**: Intellectual disability
  - **C**: Cutaneous neurofibromas
  - **L**: Lisch nodules (pigmented iris hamartomas)
  - **O**: Optic gliomas
  - **P**: Pheochromocytomas
  - **S**: Seizures
  - **S**: focal neurologic Signs (often from meningioma)
  - bone lesions (e.g., sphenoid dysplasia)



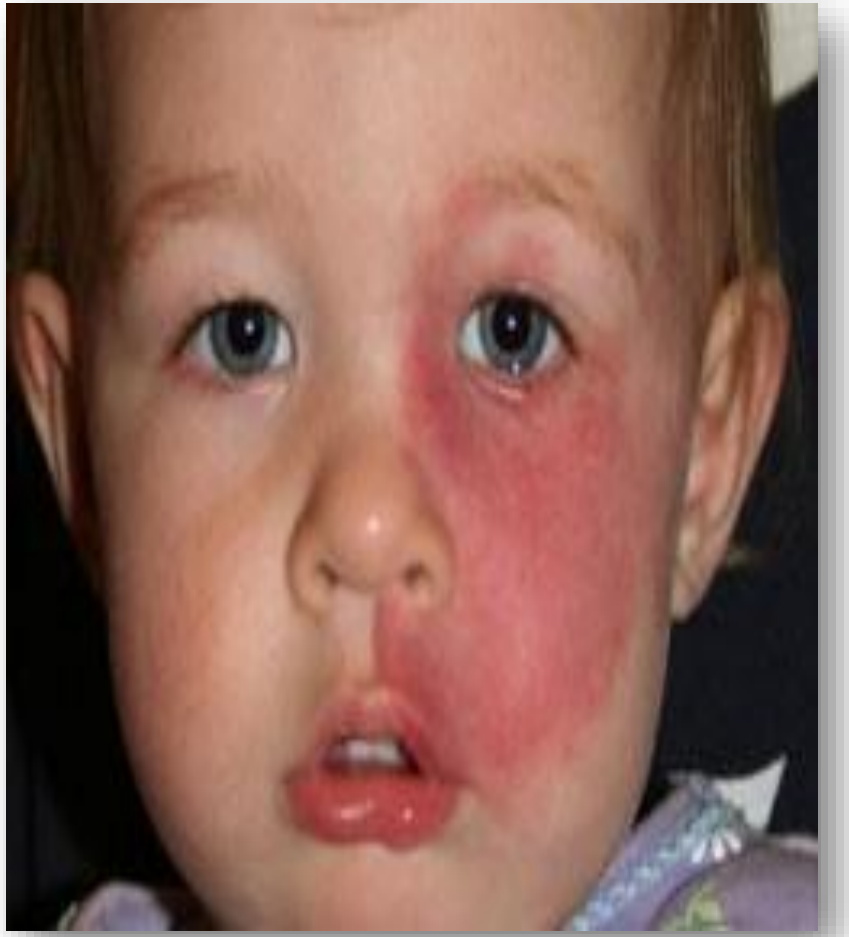
Café-au-lait spots

von Hippel-Lindau disease

- The characteristics of von Hippel-Lindau disease may be remembered with the mnemonic "**HARP**":
  - **H**: Hemangioblastomas in retina, brain stem, cerebellum, spine
  - **A**: Angiomatosis (e.g., cavernous hemangiomas in skin, mucosa, organs)
  - **R**: bilateral Renal cell carcinoma
  - **P**: Pheochromocytoma

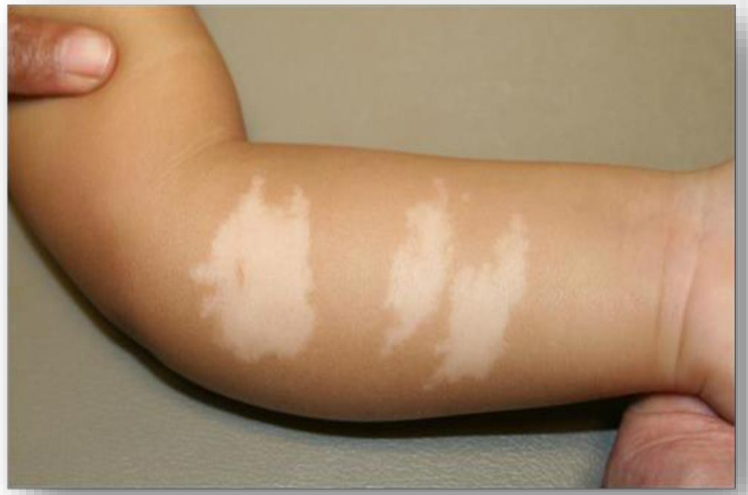
Sturge-Weber syndrome

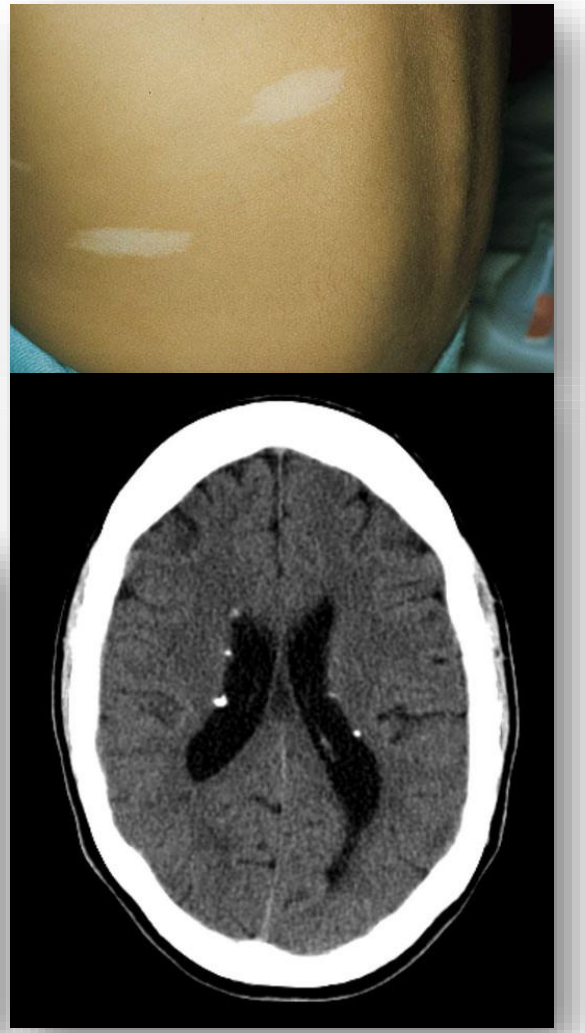
- **What is the cause ?**
  - Sturge-Weber syndrome
- **What head CT findings are associated with this disease ?**
  - Tram-track calcification
- **What eye finding is associated with this disease ?**
  - Glaucoma



Child with seizures with skin findings

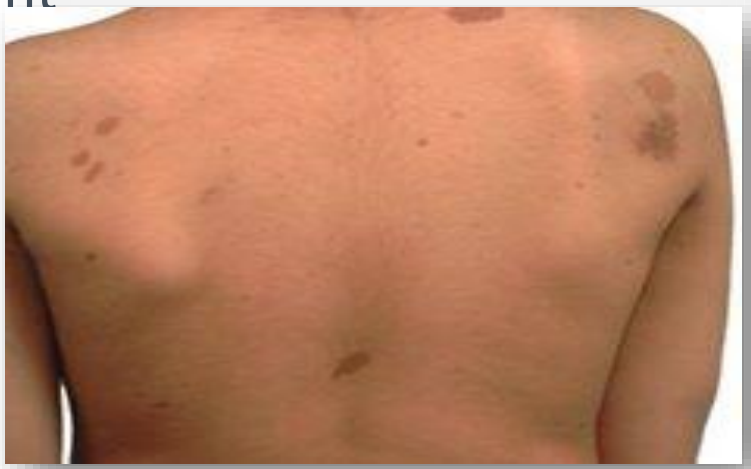
- **What is the name of this skin lesion ?**
  - Ash-leaf spots
- **What are the CT findings ?**
  - Calcification
- **What is your diagnosis ?**
  - Tuberous sclerosis

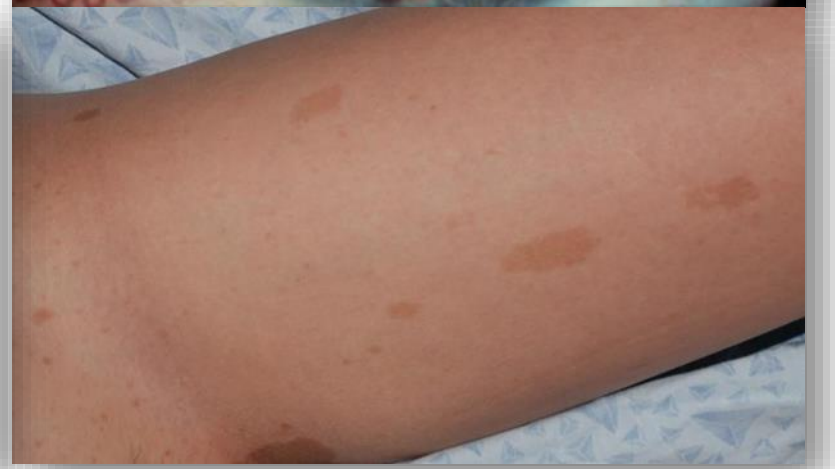




Neurofibromatosis

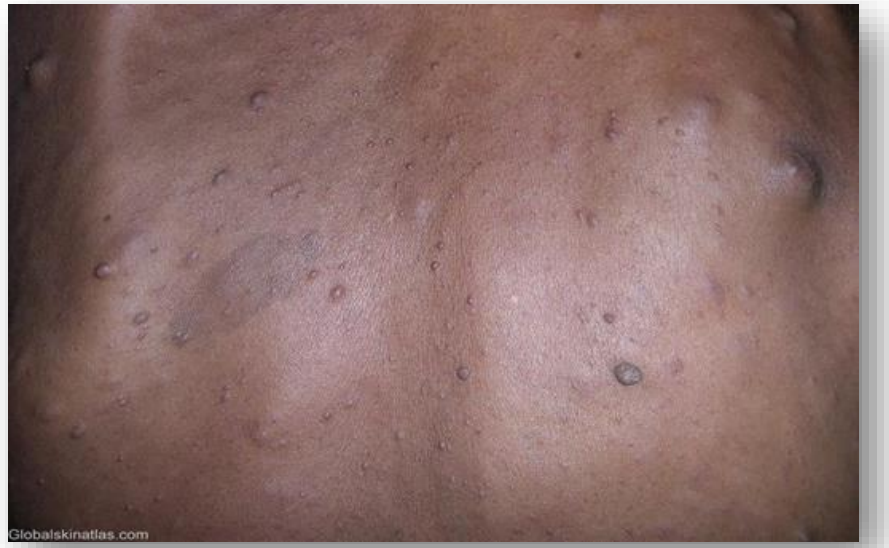
- **What is the name of this skin lesion ?**
  - Café Au Lait Spot
- **What is your deferential diagnosis ?**
  - Neurofibromatosis 1
- **What's the mode of inheritance?**
  - Autosomal dominant





Neurofibromatosis

- **Mention 2 findings seen in the picture:**
  - Café Au Lait Spot
  - Cutaneous neurofibromas
- **What is your diagnosis ?**
  - Neurofibromatosis

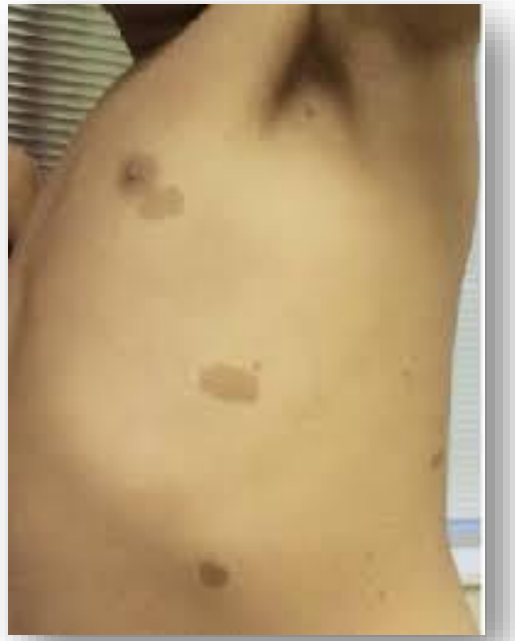


Neurofibromatosis

- 7-year-old girl presented with developed breast and blindness, skin pigmentation

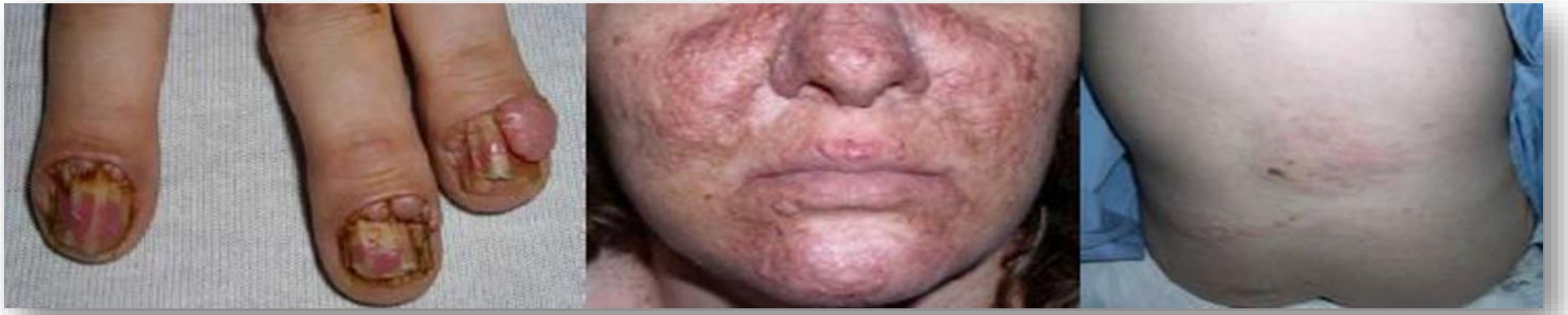
- **What is your diagnosis ?**
  - Neurofibromatosis 1

- **What is the cause of precious thelarche ?**
  - Organic/central brain lesion leads to premature secretion of GnRH or hypothalamus-pitutry-adrenal axis activation



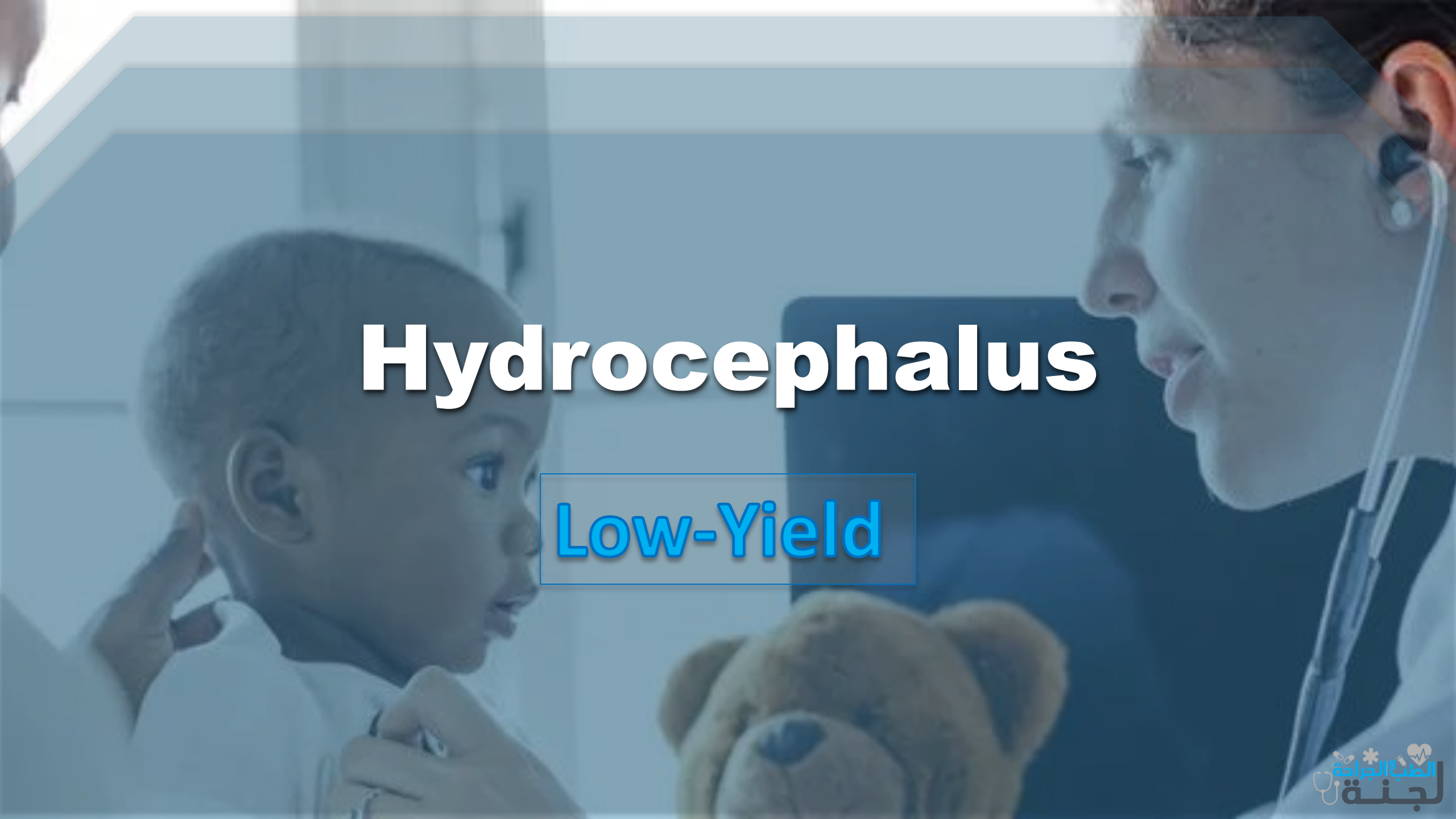
Mentally retarded patient, his brother has same condition

- **Mention 2 signs.**
  - Ungual fibromas, Adenoma sebaceum, Shagreen patch
- **What's the Diagnosis?**
  - Tuberous sclerosis.
- **Name 2 other investigations you will order to look for more signs.**
  - Contrast brain CT/MRI, kidney CT, echocardiography, ECG, EEG



Hydrocephalus

Low-Yield



Hydrocephalus

Symptoms	Physical Exam Findings
Poor feeding	Tense and bulging fontanelle
Irritability	Prominent scalp veins
Decreased activity	Widely spaced cranial sutures
Vomiting	Rapidly increasing head circumference

Hydrocephalus

❖**What is the name of this sign ?**

- Sunset eyes (setting sun sign)

❖**What is the next examination ?**

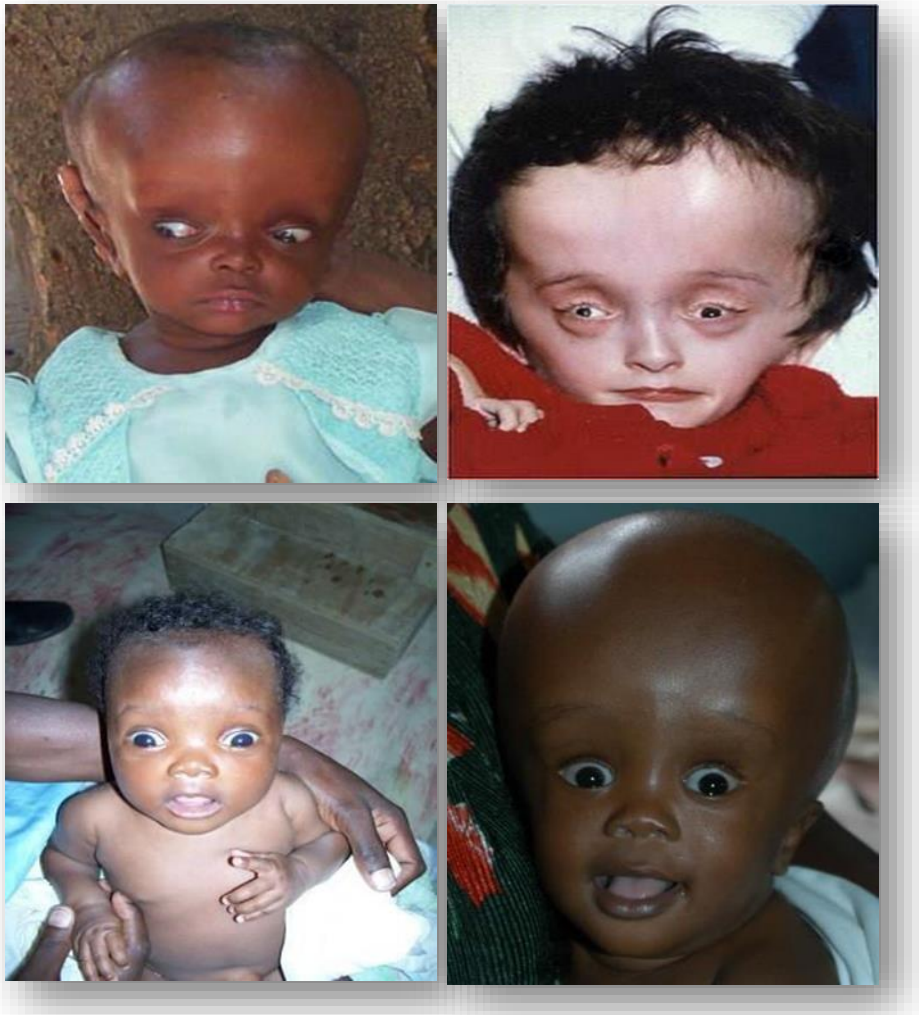
- Head circumference

❖**The child is having which of the following ?**

a. Exophthalmia
b. Hydrocephalus
c. 3rd cranial nerve palsy
d. Horner syndrome
e. 6th nerve palsy

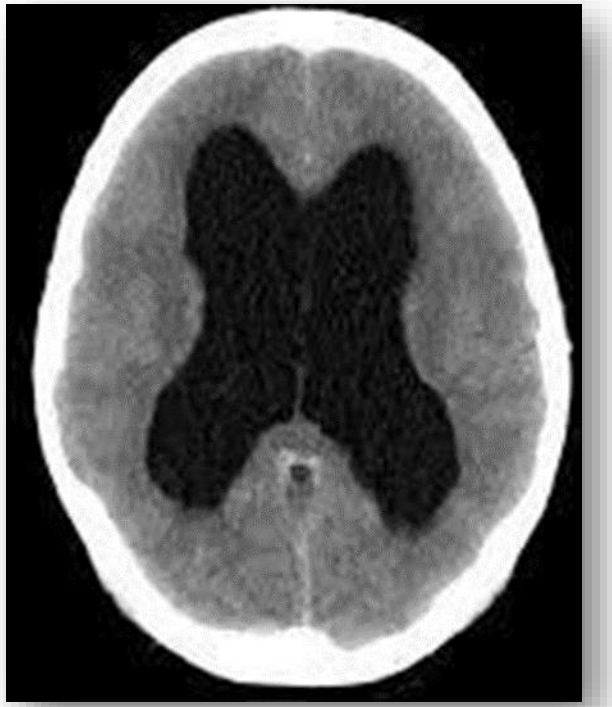
Hydrocephalus

- **What is your Diagnosis ?**
  - Hydrocephalus
- **Mention 2 signs.**
  - Increased head circumference
  - Bulging anterior fontanelle
  - Sun set eyes



CT scan of a child, his head circumference is at 97th

- **Mention 2 signs**
  - Widening of the ventricles (ventriculomegaly)
  - Effacement of sulci
- **Mention 2 symptoms**
  - Headache & Projectile vomiting

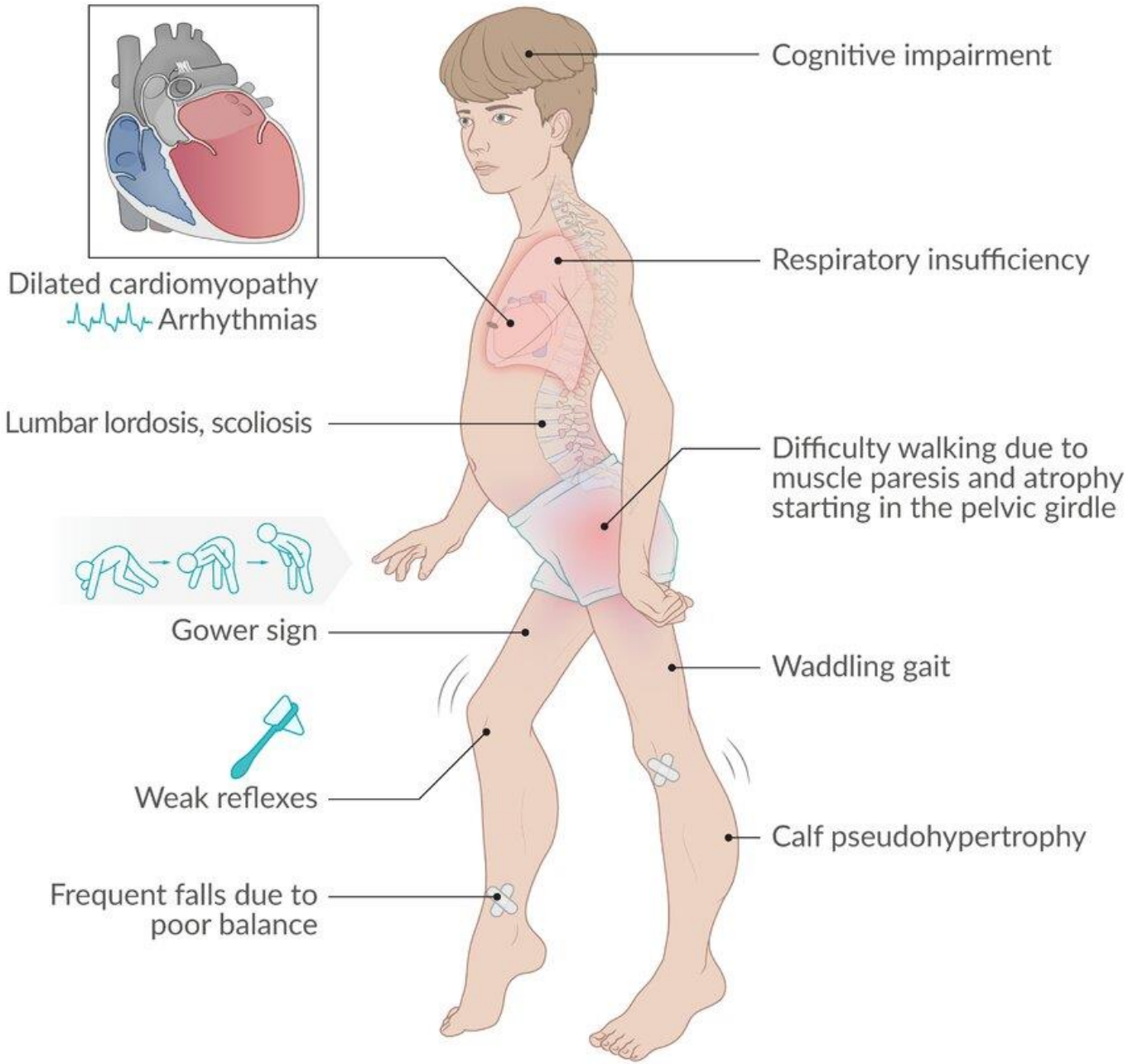


Muscular dystrophy

Low-Yield

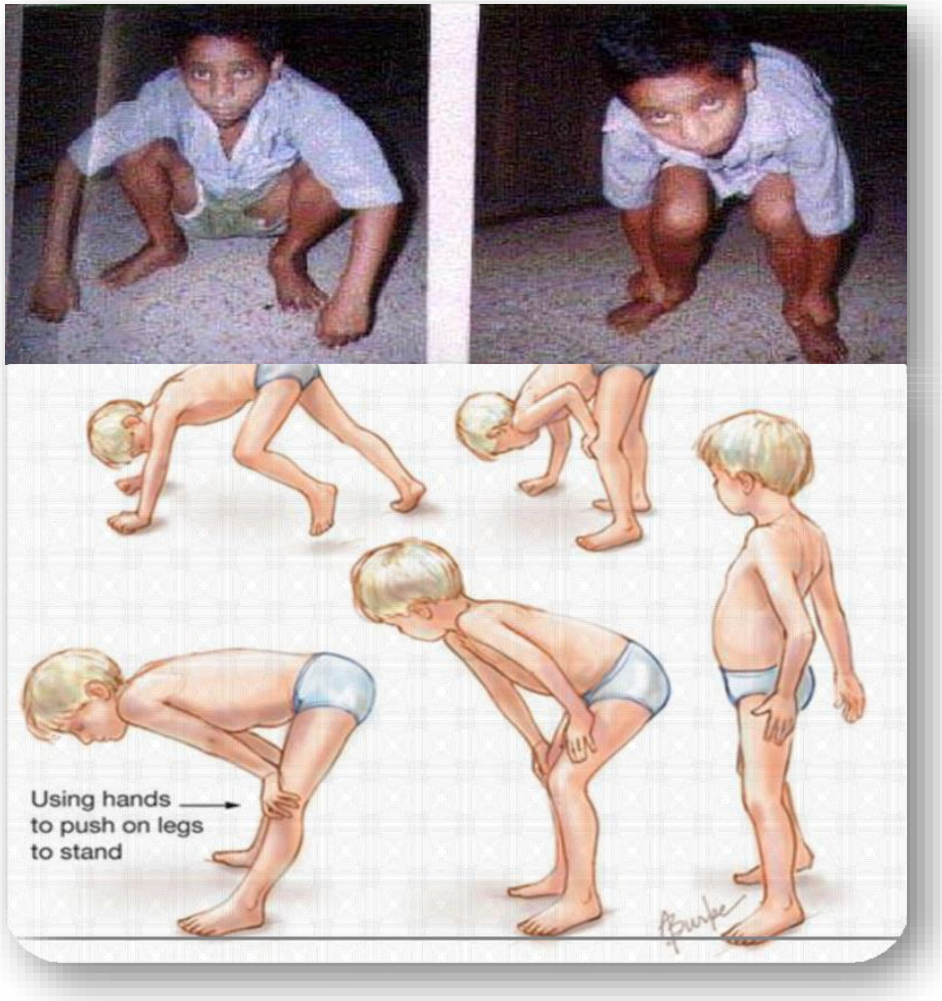
إضافي

	Duchenne muscular dystrophy	Becker muscular dystrophy
Etiology	**X-linked recessive**, dystrophin gene mutation	
Dystrophin protein	**Absent**	**Reduced**
Age of onset	2–5 years	> 15 years
Symptoms	Rapid progression, inability to walk by approx. 12 years	Less severe, slower progression, cardiac involvement is more common
Diagnostics	↑↑ **Creatine kinase,** ↑ serum aldolase, genetic analysis (confirmatory test), muscle biopsy	
Treatment	Supportive, glucocorticoids	
Life expectancy	~ 30 years	~ 40–50 years



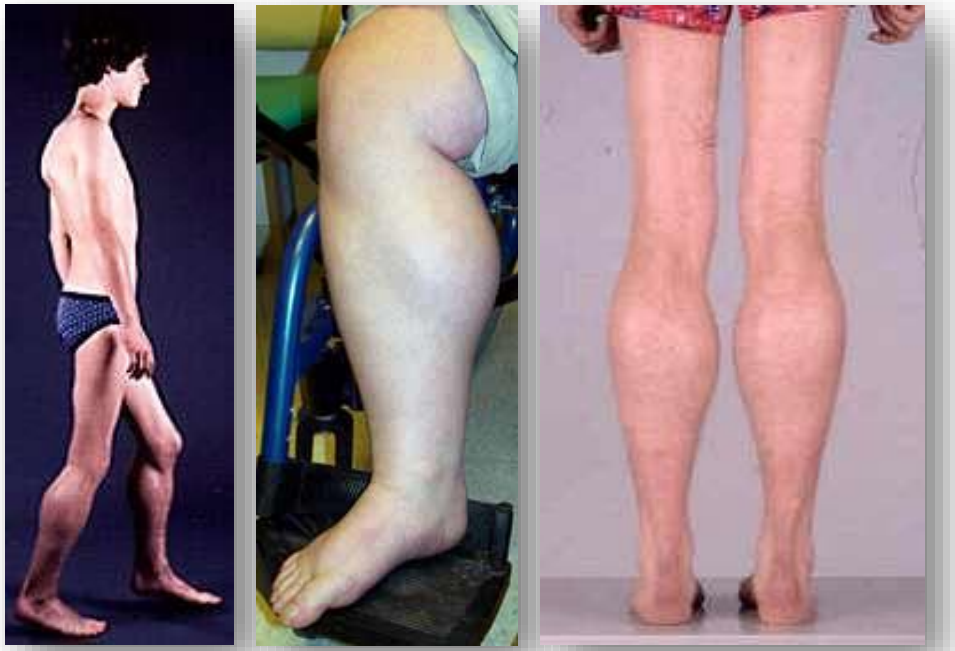
Gower sign

- **What is this sign ?**
  - Gower sign
- **Mention 3 causes ?**
  - Duchenne muscular dystrophy
  - Becker muscular dystrophy
  - Myotonic muscular dystrophy
  - juvenile dermatomyositis
- **What group of muscles are affected ?**
  - Proximal muscle wasting
- **What is the pattern of inheritance ?**
  - X-linked recessive

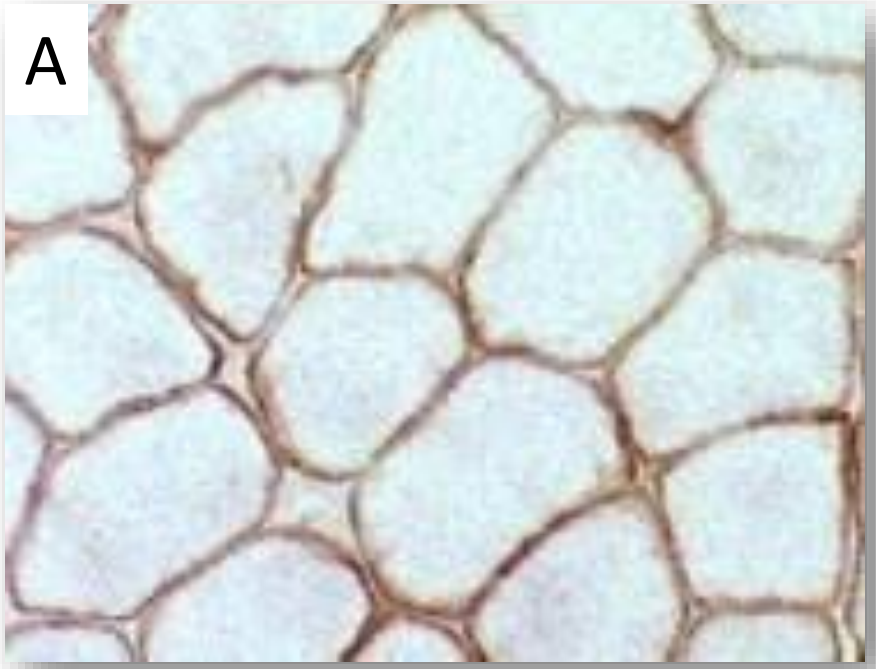


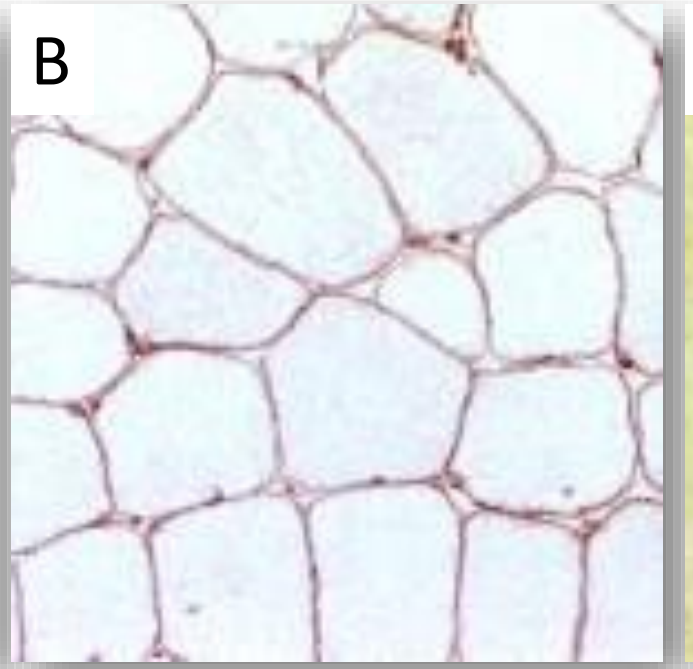
Write three differential diagnoses of this condition

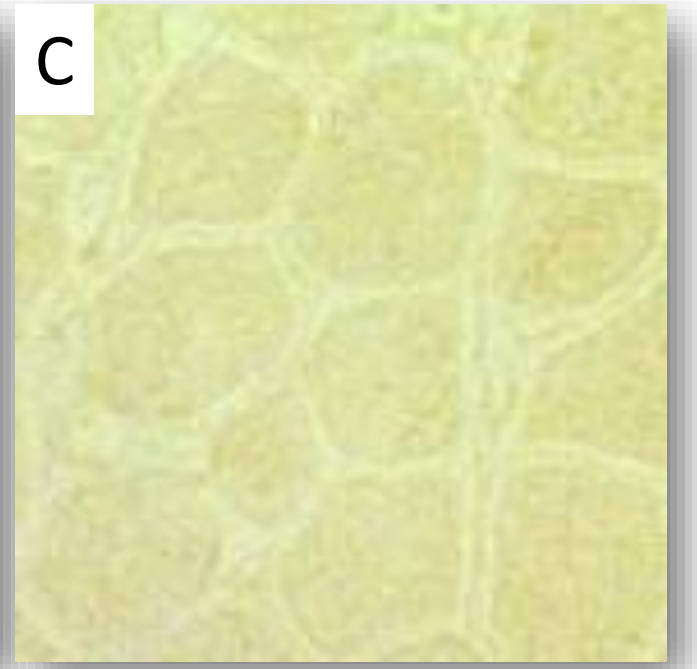
1. Duchene muscular dystrophy
2. Becker muscular dystrophy
3. Limb girdle muscular dystrophy



A is normal, what is the condition in B & C







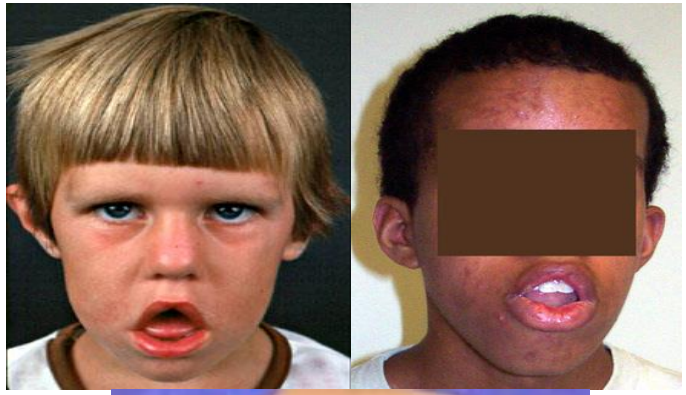
Normal

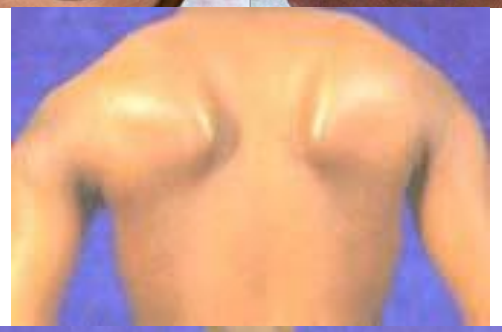
Becker

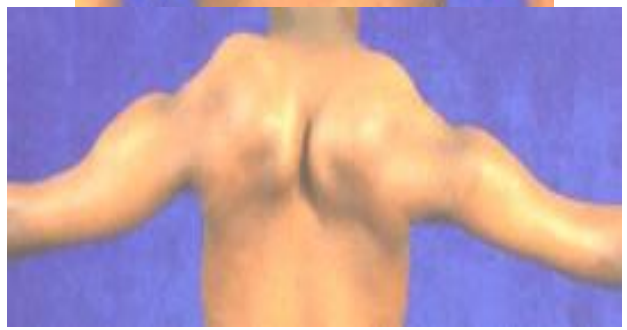
Duchene

Myotonic muscular dystrophy

- **What dysmorphic features are seen in the pictures ?**
  - Facial wasting
  - Inverted V shape upper lip
  - Thin cheeks, scalloped, concave tempo ales
  - Narrow head, cleft lip
  - Wasted sternocleidomastoid muscles
  - Neck thin, long, cylindrical contour
  - Scapular winging



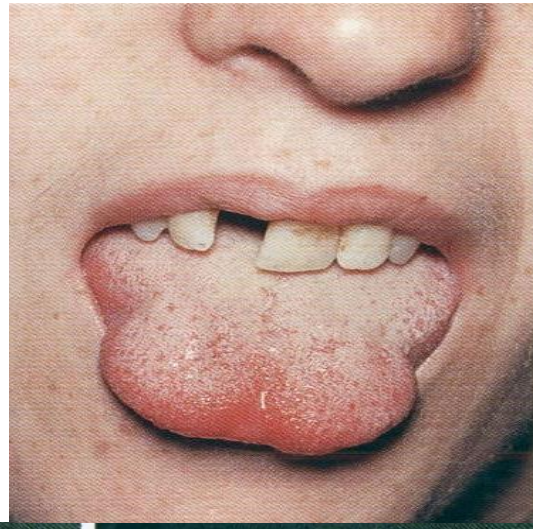




Myotonia

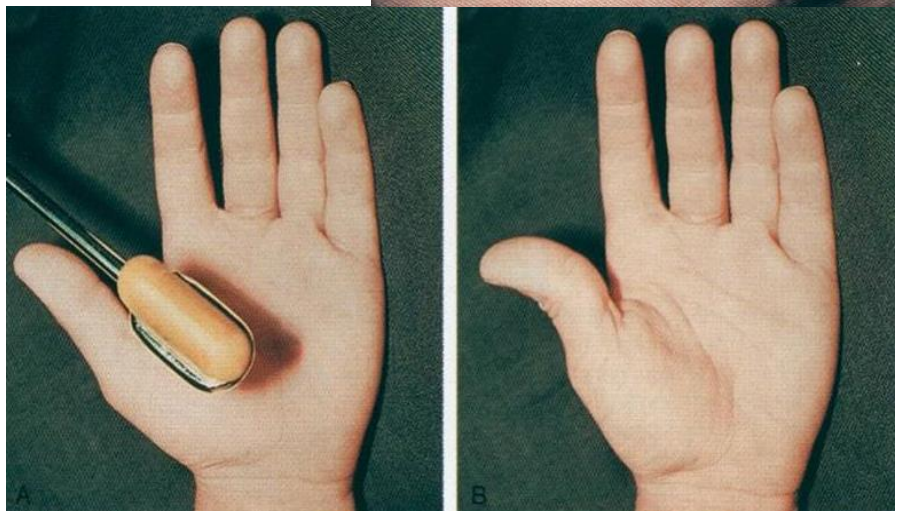
❖Characteristics

- Slow relaxation of muscle after contraction
- Not evident before 5 years
- Characteristic features shared by muscular dystrophies
- It is not painful
- No correlation with weakness



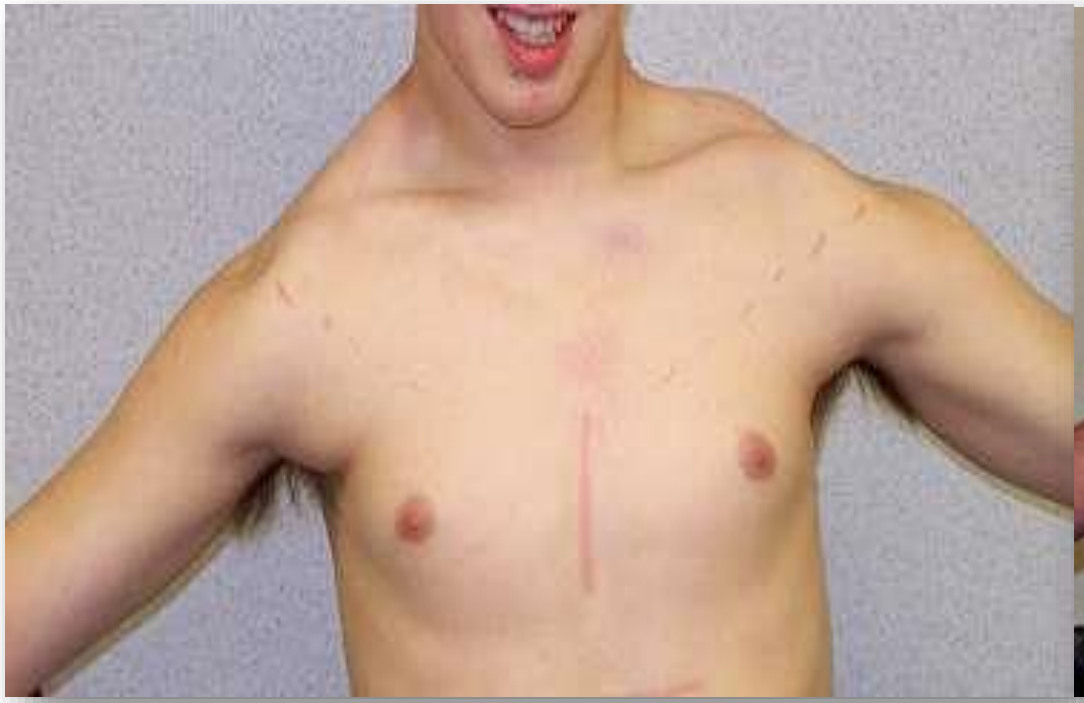
❖Examination for myotonia

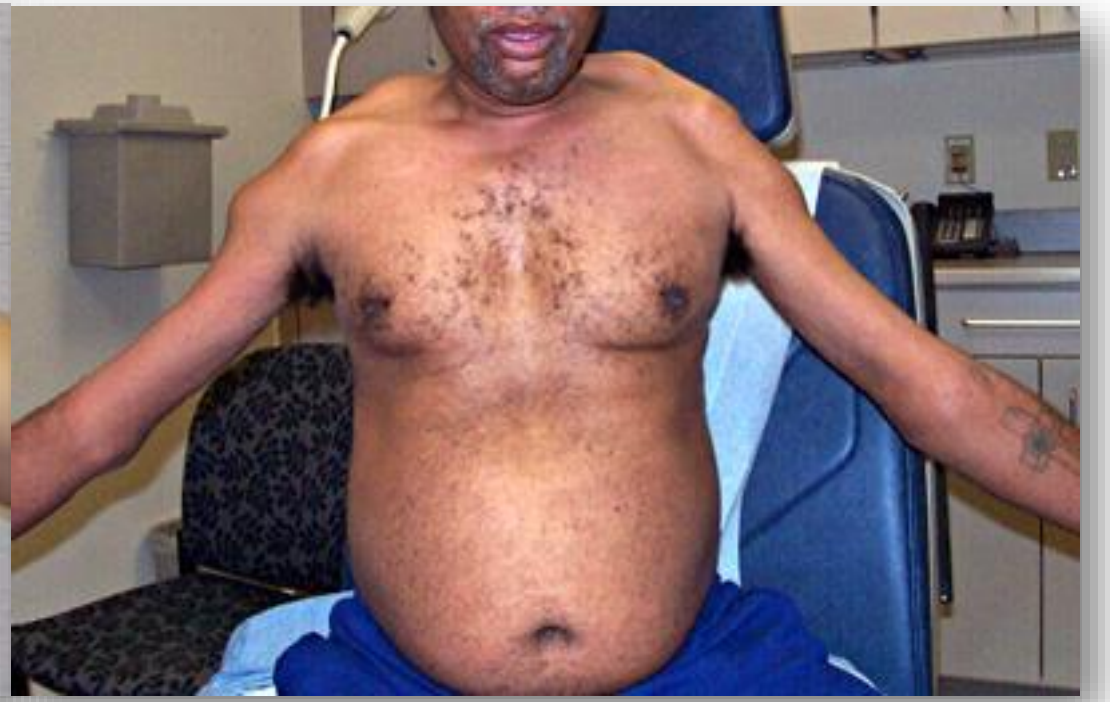
- Make fist
- Striking thenar eminence with hammer
- Pressing tongue with blade



Shoulder muscles wasting

- **What findings are seen in the pictures ?**
  - Shoulder muscles wasting; not a specific sign for a particular dystrophy





Cerebral palsy

Low-Yield

Cerebral Palsy

- **Definition**: persistent (not progressive) disorder of movements and/or posture caused by a lesion to the immature brain
- **Diagnosis made clinically by history and physical exam**
  - The prenatal and perinatal history are very important → Any problems in these augment the diagnosis
- **Prenatal causes 75%**:
  - Rubella, CMV, Toxoplasmosis, Brain malformation, Genetic factors
- **Perinatal causes 10%**:
  - Most important cause is hypoxic encephalopathy
- **Postnatal causes 15%**:
  - Hyperbilirubinemia, Bacterial meningitis, Viral encephalitis, Shaken baby syndrome

Classification

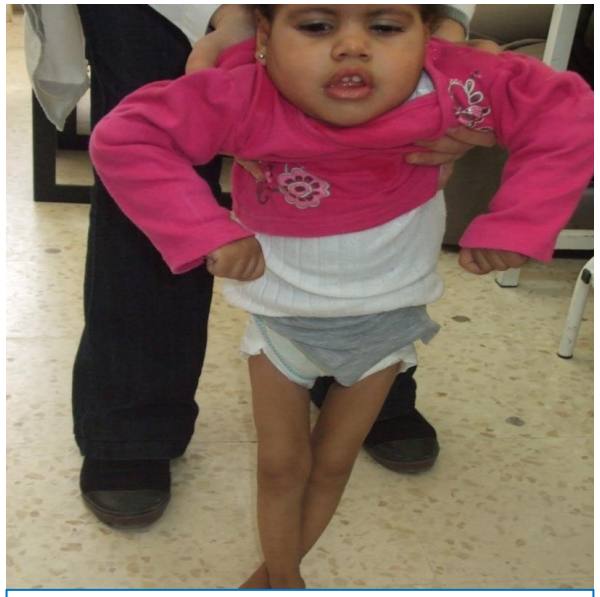
- **Spastic**
  - **Hemiplegia**: Arm bent, Tip toe walking, Hand preference at early age
  - **Diplegia**: Scissoring posture
  - **Quadriplegia**: Involve all extremities, associated with seizures, speech affected
- **Choreoathetoid**
  - Tongue thrust drooling feeding difficulties
  - Speech affected
  - Seizures are common
  - Intellect is preserved in many patient
  - Associated with asphyxia and hyperbilirubinemia
- **Mixed**
  - Presence of athetoid movement in one limb and hemiplegia in the other or presence of ataxia and spasticity
- **Ataxic**: Hypotonia then spasticity

إضافي

Spastic cerebral palsy

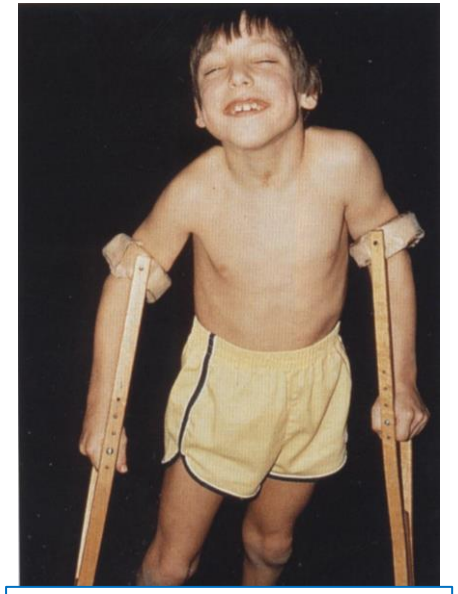
Spastic hemiplegia

- 1 Side completely normal
- Other side:
  - Arm bent spastic or floppy, often of no use
  - Walks on tiptoe



Spastic diplegia

- Bilateral spasticity of legs greater than arms
- Scissoring posture
- Delayed walking tip toe walking

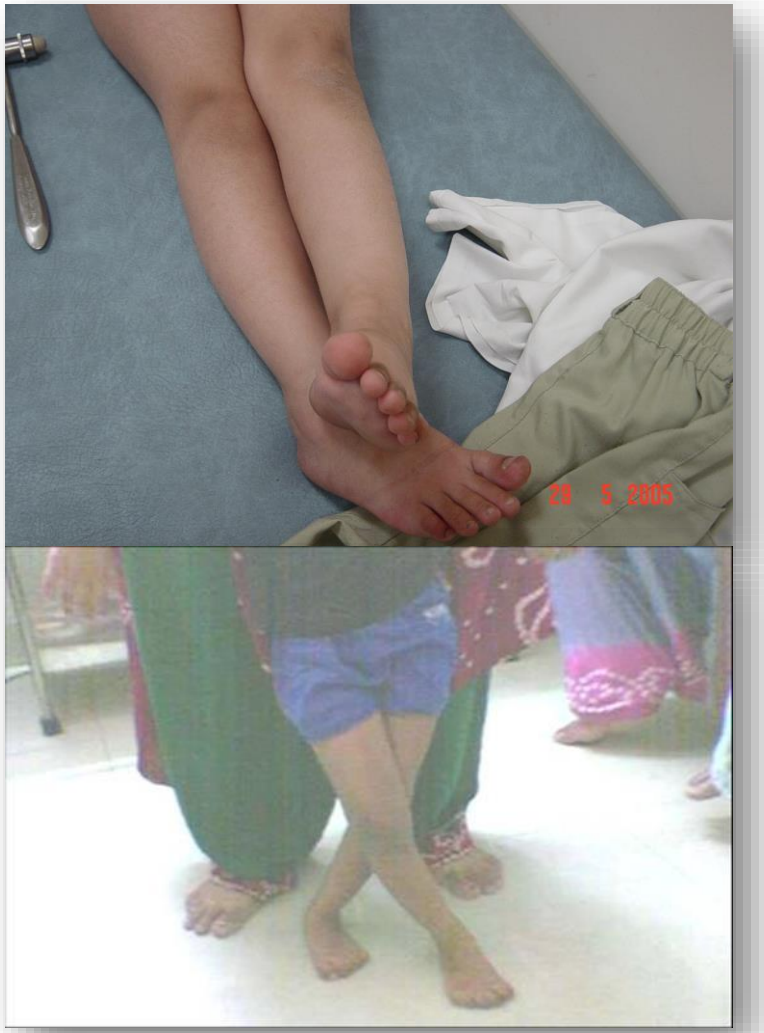


Spastic quadriplegia

- Most severe form
- Involve all extremities

Cerebral palsy

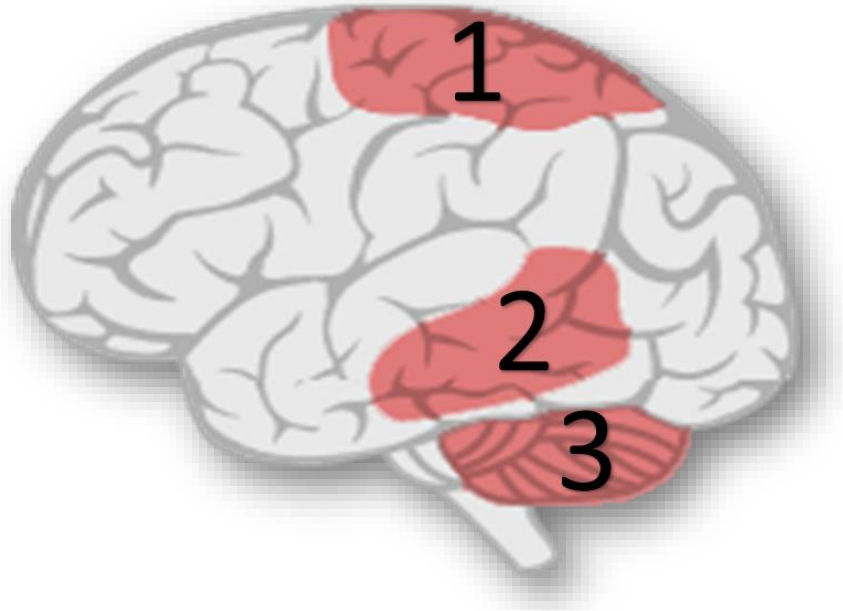
- **What's the sign called ?**
  - Scissoring sign
- **Give one disease you can see this sign?**
  - Spastic CP



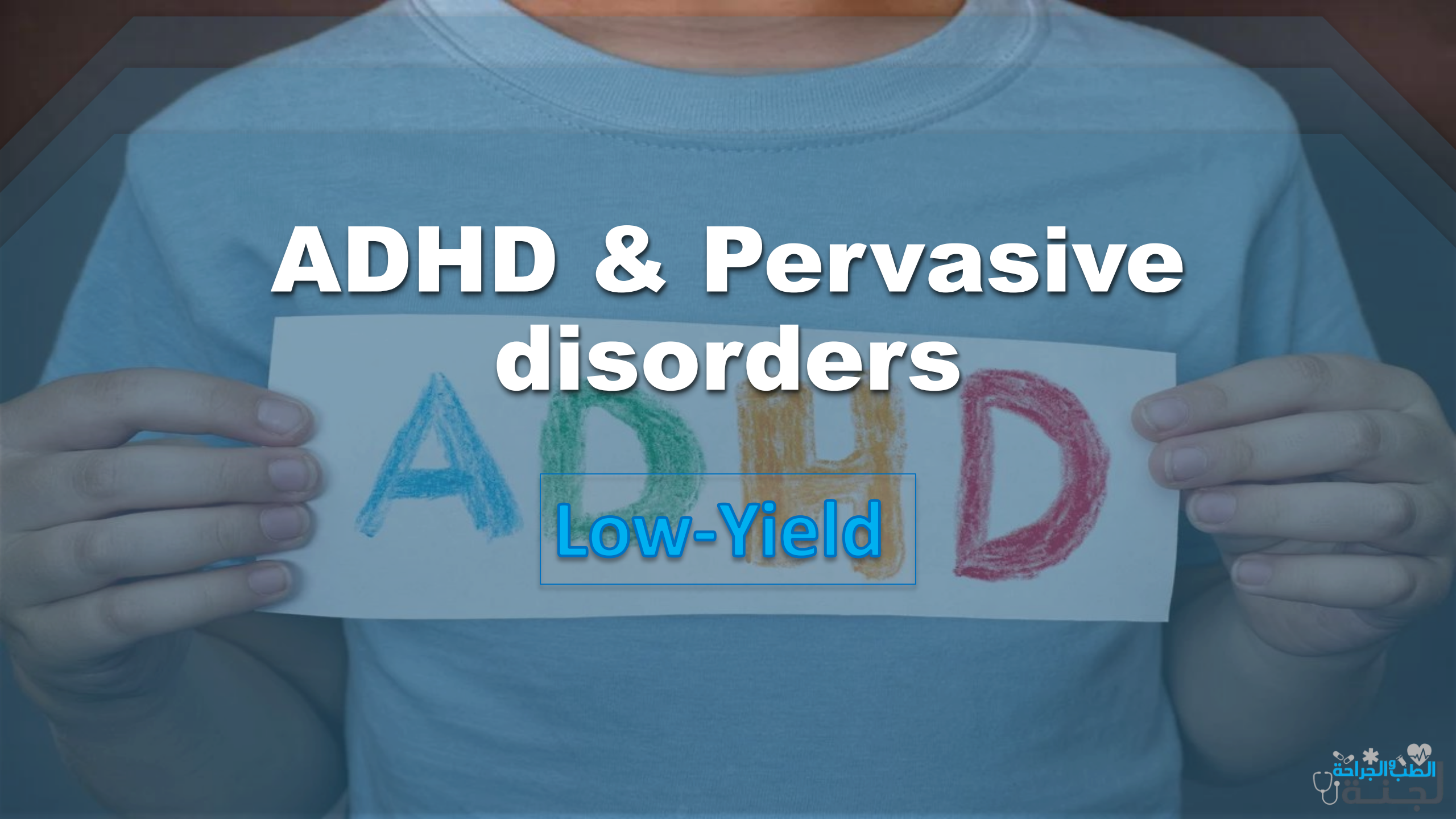
Cerebral palsy

❖ **What is the type of cerebral palsy for each area ?**

- 1 → Spastic palsy
- 2 → Dyskinetic (choreoathetoid) palsy
- 3 → Ataxic palsy
- 1 + 2 + 3 → Mixed cerebral palsy



ADHD & Pervasive disorders



Low-Yield

ADHD

- 5-year-old patient with autism and ADHD
- **What is the cause ?**
  - Fragile-X syndrome

- Autism spectrum disorder may be associated fragile X syndrome and double Y males (XYY)

إضافي

Fragile X syndrome

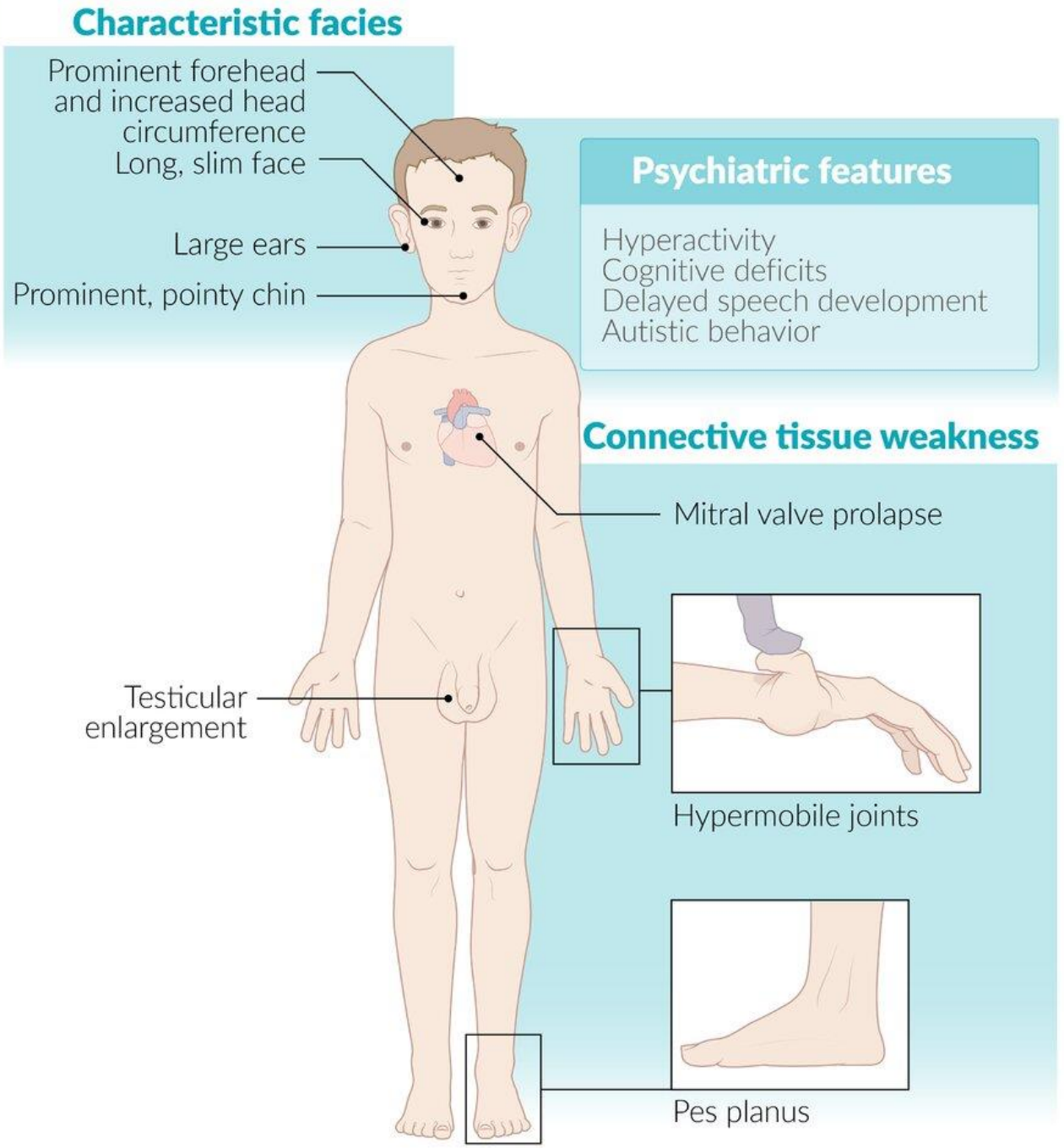
Epidemiology
Approx. 1 in 8,000 females and 1 in 4000 males
Second most common cause of congenital cognitive disorders after trisomy 21

Etiology
Mutation of the FMR1 gene
(CGG trinucleotide repeat expansion)
X-linked inheritance

Complications
Epilepsy

Note
Milder manifestations in heterozygous female individuals
(mostly neuropsychiatric disorders, rarely cognitive deficits)

Life expectancy
Typically normal



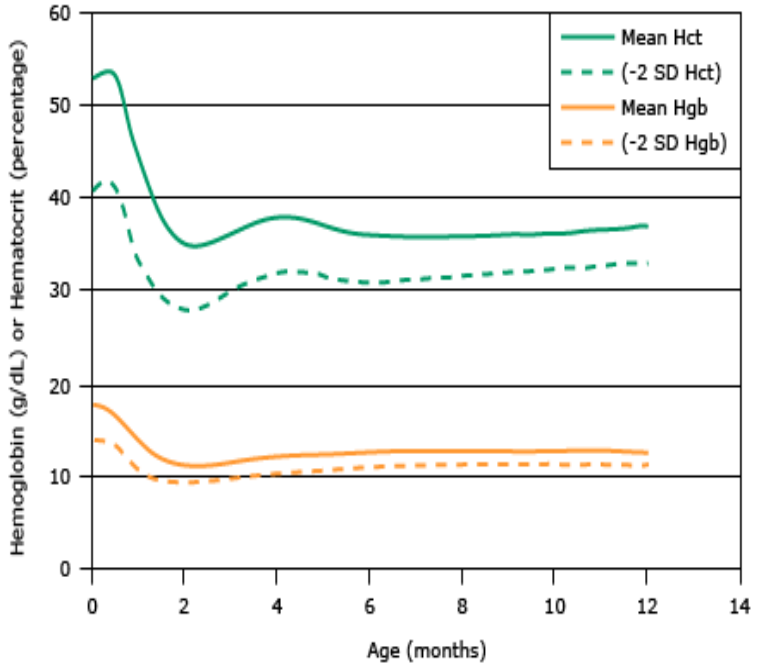
Hematoncology


Anemia in pediatrics
High-Yield

Physiologic Anemia Of Infancy

	Preterm	Full term
Timing	6-8 weeks	8-12 weeks
Hgb levels	7-9 g/dL	9-11 g/dL
Treatment	No treatment is needed	

Normal values for hematocrit and hemoglobin during the first year of life in healthy term infants



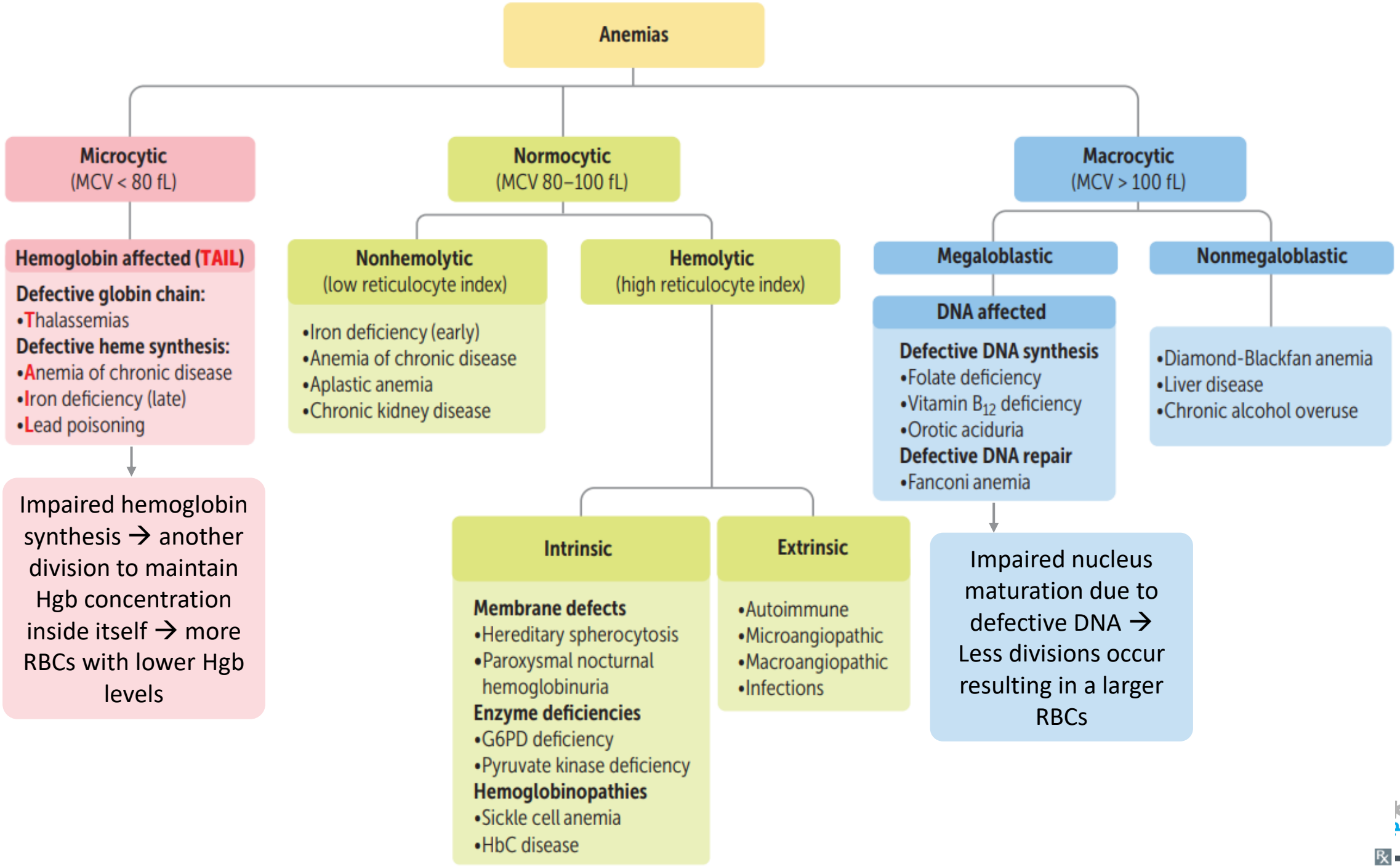
Physiologic Anemia Of Infancy

- Pathologic anemia in newborns and young infants is distinguished from physiologic anemia by any of the following
  - Anemia (HGB <13.5 g/dL) within the first month of life
  - Anemia with lower HGB level than is typically seen with physiologic anemia (i.e., <9 g/dL)
  - Signs of hemolysis (e.g., jaundice, scleral icterus, or dark urine) or symptoms of anemia (e.g., irritability or poor feeding

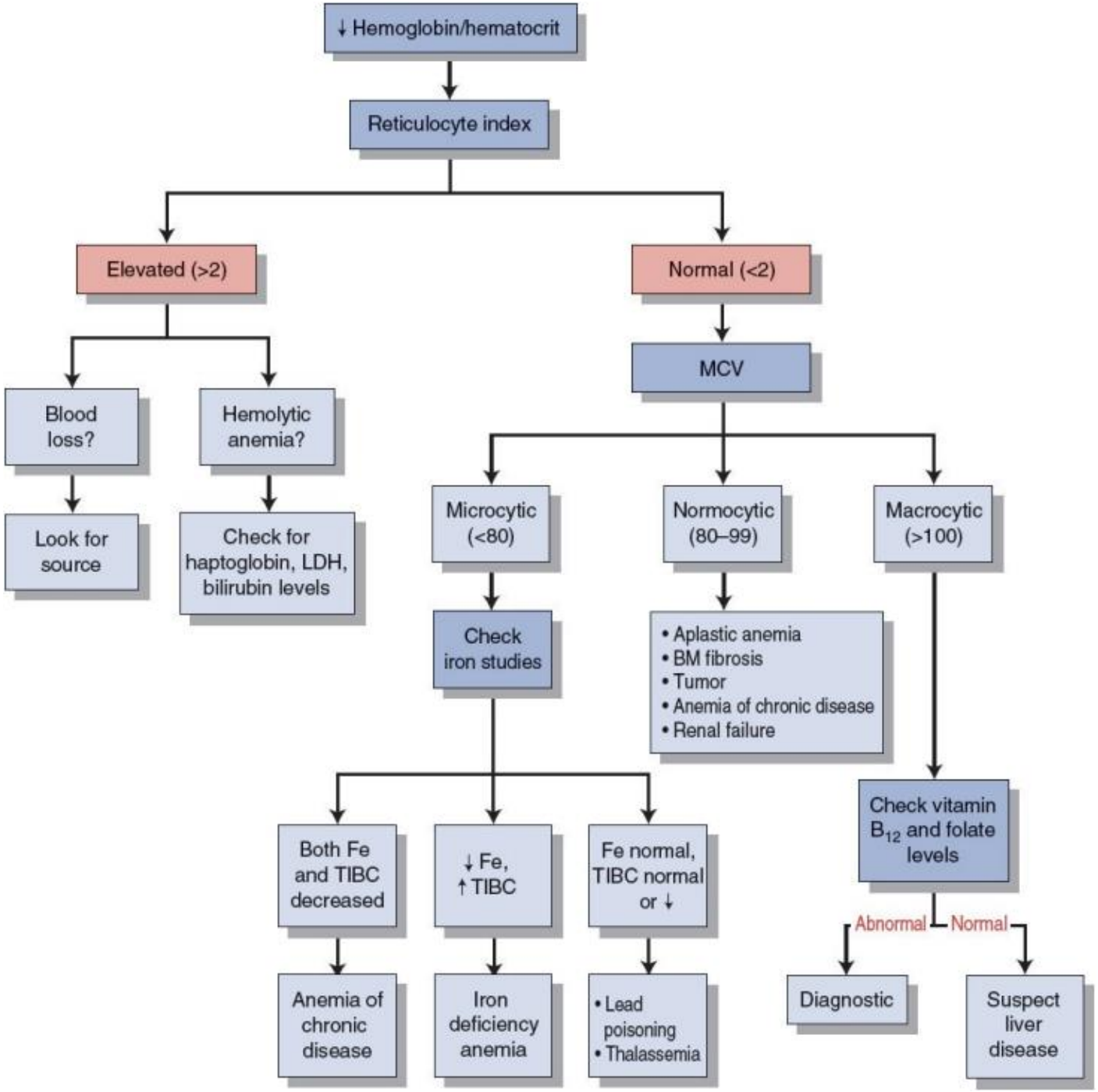
Terminology

- **Hematocrit (HCT)**: Is the fractional volume of a whole blood sample occupied by RBCs, expressed as a percentage. (قديش نسبة خلايا الدم الحمراء في الدم)
- **Hemoglobin (HGB)**: This is a measure of the concentration of the RBC pigment HGB in whole blood, expressed as grams per 100 mL (dL) of whole blood. (قديش نسبة الهيموجلوبين في الدم)
- **Mean corpuscular hemoglobin concentration (MCHC)**: is a calculated index (MCHC = HGB/HCT) yielding a value of grams of HGB per 100 mL of RBC. (قديش نسبة الهيموجلوبين في خلايا الدم الحمراء)
- **Mean corpuscular volume (MCV)**: Represents the mean value (in femtoliters [fL]) of the volume of individual RBCs in the blood sample.
- **Red cell distribution width (RDW)**: The RDW is a quantitative measure of the variability of RBC sizes in the sample (anisocytosis).

شرح


Anemias
Microcytic
(MCV < 80 fL)
Normocytic
(MCV 80–100 fL)
Macrocytic
(MCV > 100 fL)
Hemoglobin affected (TAIL)
Defective globin chain:
•Thalassemias
Defective heme synthesis:
•Anemia of chronic disease
•Iron deficiency (late)
•Lead poisoning
Impaired hemoglobin synthesis → another division to maintain Hgb concentration inside itself → more RBCs with lower Hgb levels
Nonhemolytic
(low reticulocyte index)
•Iron deficiency (early)
•Anemia of chronic disease
•Aplastic anemia
•Chronic kidney disease
Hemolytic
(high reticulocyte index)
Intrinsic
Membrane defects
•Hereditary spherocytosis
•Paroxysmal nocturnal hemoglobinuria
Enzyme deficiencies
•G6PD deficiency
•Pyruvate kinase deficiency
Hemoglobinopathies
•Sickle cell anemia
•HbC disease
Extrinsic
•Autoimmune
•Microangiopathic
•Macroangiopathic
•Infections
Megaloblastic
Nonmegaloblastic
DNA affected
Defective DNA synthesis
•Folate deficiency
•Vitamin B12 deficiency
•Orotic aciduria
Defective DNA repair
•Fanconi anemia
•Diamond-Blackfan anemia
•Liver disease
•Chronic alcohol overuse
Impaired nucleus maturation due to defective DNA → Less divisions occur resulting in a larger RBCs

Evaluation of anemia



Differential diagnosis of neonatal anemia

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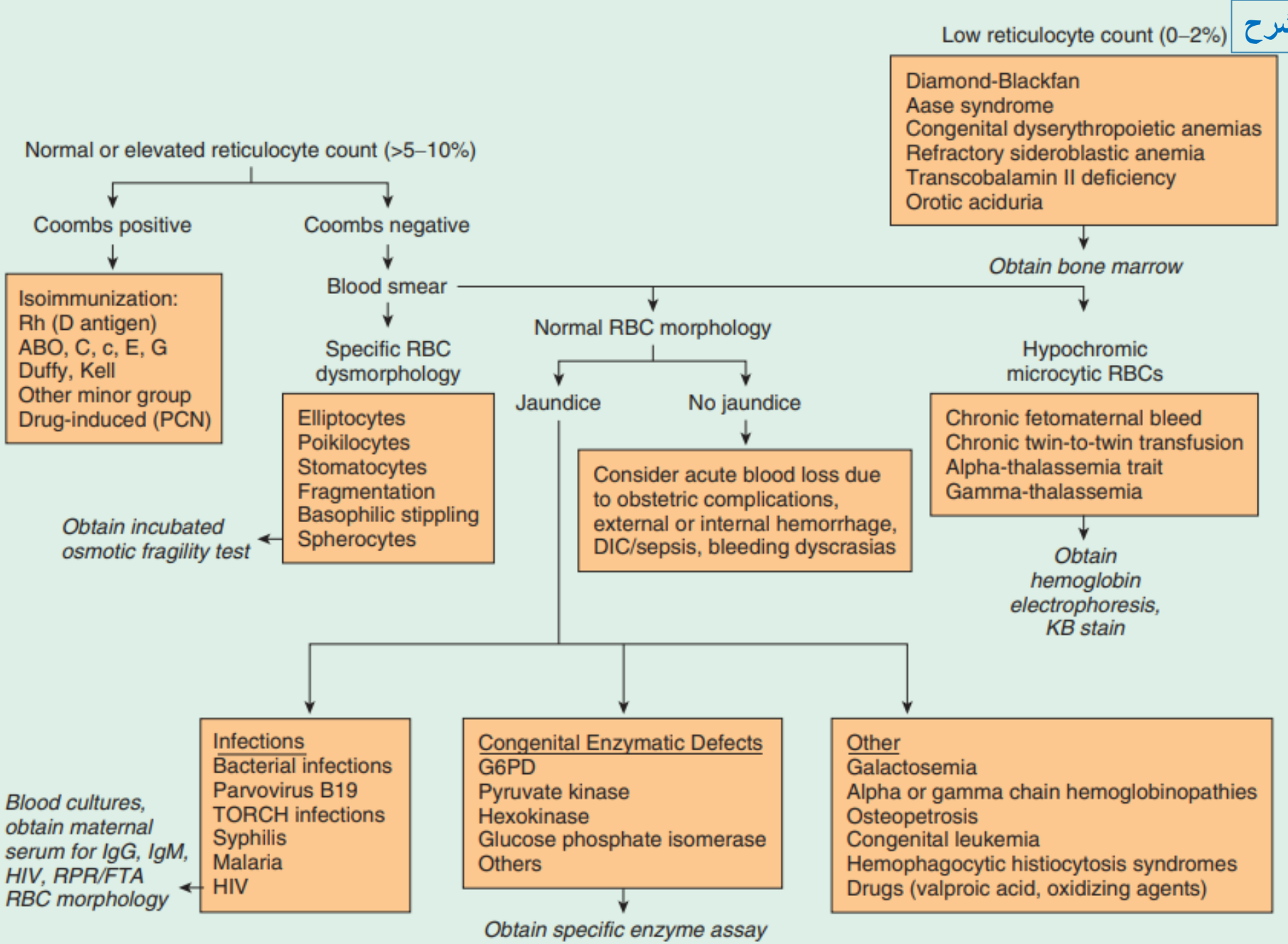
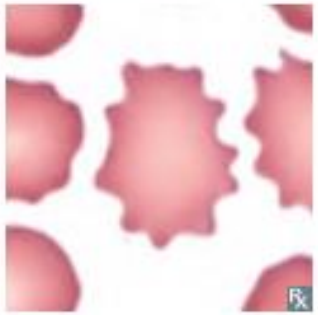
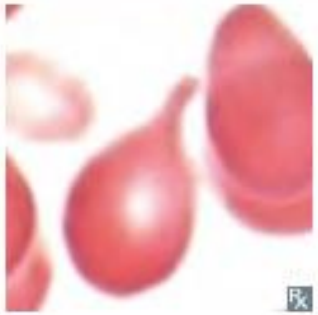


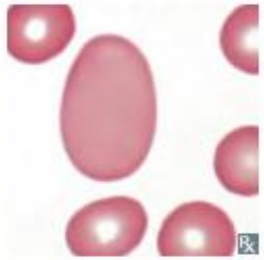
RBC morphology

TYPE	EXAMPLE	ASSOCIATED PATHOLOGY	NOTES
Acanthocytes ("spur cells")		Liver disease, abetalipoproteinemia, vitamin E deficiency	Projections of varying size at irregular intervals (acanthocytes are asymmetric).
Echinocytes ("burr cells")		Liver disease, ESRD, pyruvate kinase deficiency	Smaller and more uniform projections than acanthocytes (echinocytes are even).
Dacrocytes ("teardrop cells")		Bone marrow infiltration (eg, myelofibrosis)	RBC "sheds a tear" because it's mechanically squeezed out of its home in the bone marrow

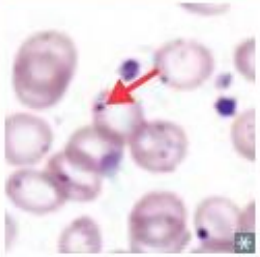
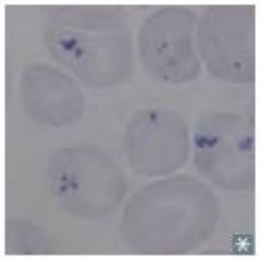
RBC morphology

TYPE	EXAMPLE	ASSOCIATED PATHOLOGY	NOTES
Schistocytes ("helmet" cells)		MAHAs (eg, DIC, TTP/HUS, HELLP syndrome), mechanical hemolysis (eg, heart valve prosthesis)	Fragmented RBCs
Degmacytes ("bite cells")		G6PD deficiency	Due to removal of Heinz bodies by splenic macrophages (they "deg" them out of/bite them off of RBCs)
Elliptocytes		Hereditary elliptocytosis	Caused by mutation in genes encoding RBC membrane proteins (eg, spectrin)

RBC morphology

TYPE	EXAMPLE	ASSOCIATED PATHOLOGY	NOTES
Spherocytes		Hereditary spherocytosis, autoimmune hemolytic anemia	Small, spherical cells without central pallor ↓ surface area-to-volume ratio
Macro-ovalocytes		Megaloblastic anemia (also hypersegmented PMNs)	
Target cells		HbC disease, Asplenia, Liver disease, Thalassemia	"HALT," said the hunter to his target ↑ surface area-to-volume ratio
Sickle cells		Sickle cell anemia	Sickling occurs with low O_2 conditions (eg, high altitude, acidosis)

RBC inclusions

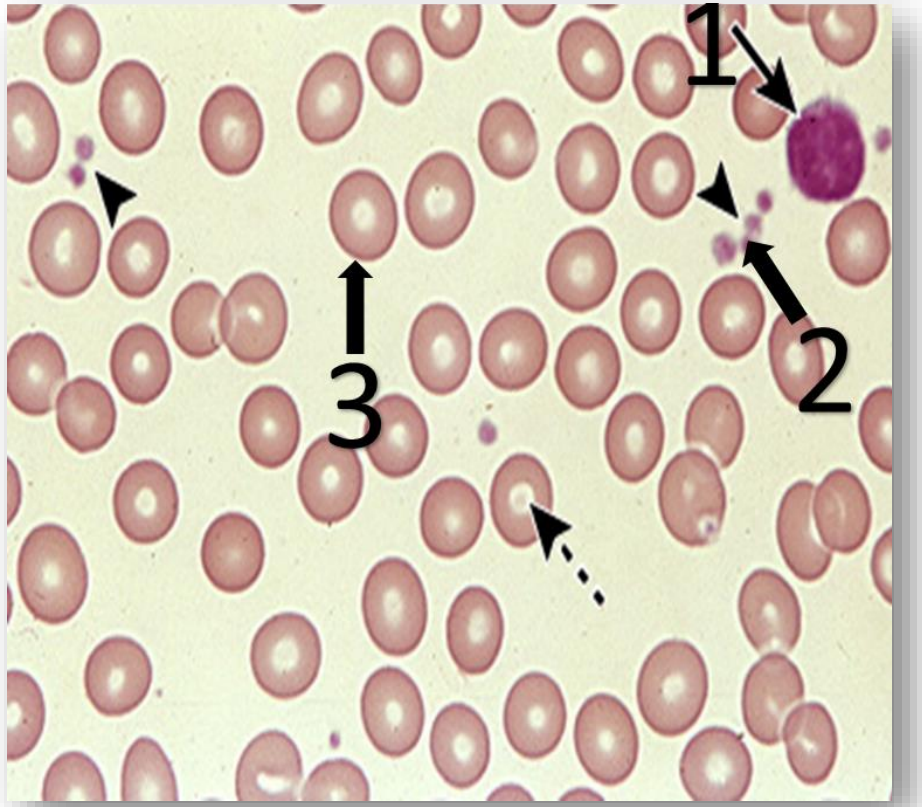
TYPE	EXAMPLE	ASSOCIATED PATHOLOGY	NOTES
Howell-Jolly bodies		Functional hyposplenia (eg, sickle cell disease), asplenia	Basophilic nuclear remnants (do not contain iron) Usually removed by splenic macrophages
Basophilic stippling		Sideroblastic anemias, thalassemias	Basophilic ribosomal precipitates (do not contain iron)
Pappenheimer bodies		Sideroblastic anemia	Basophilic granules (contain iron)
Heinz bodies		G6PD deficiency	Denatured and precipitated hemoglobin (contain iron) Phagocytic removal of Heinz bodies → bite cells Requires supravital stain (eg, crystal violet) to be visualized

NORMAL ☺

- A child came to the hematologic clinic with his family complain from bone pain

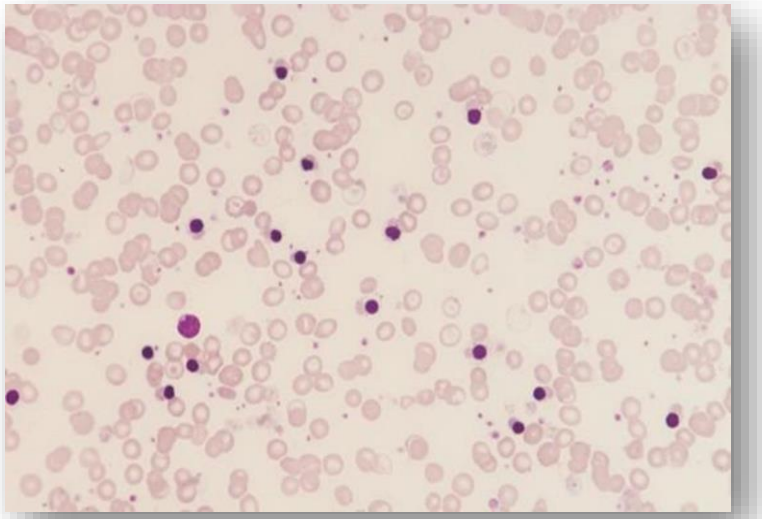
- **Is there any abnormality in this blood film, and if yes what is the diagnosis?**
  - No, it's normal blood film

- **What is the numbered cells?**
  1. Normal lymphocyte
  2. Several platelets
  3. Normal red blood cell



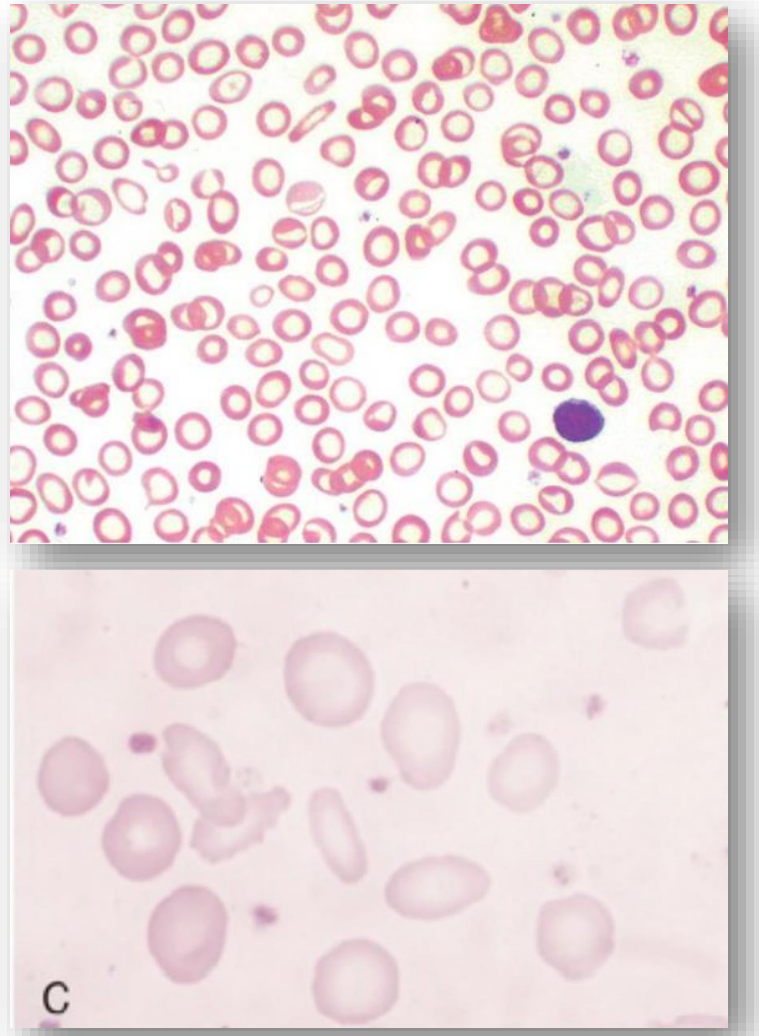
CBC shows low MCV, low MCHC, Low reticulocytes

- **What's the type of this anemia?**
  - Microcytic Hypochromic.
- **What 2 investigations you want to order?**
  - Hemoglobin Electrophoresis, Ferritin Level.



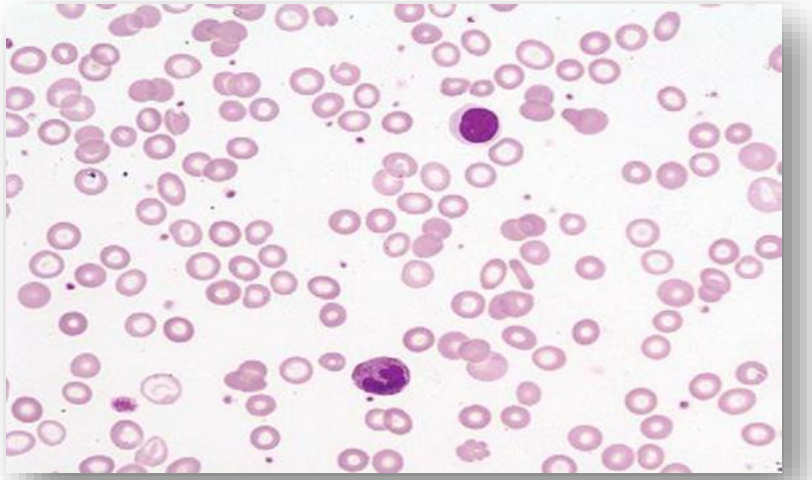
Microcytic anemia

- **What findings are seen in blood film ?**
  - Microcytic & Hypochromic RBCs.
- **What are the possible ddx ? (T.A.I.L.S)**
  - **T**halassemia
  - **A**nemia of chronic disease
  - **I**ron deficiency anemia
  - **L**ead poisoning
  - **S**ideroblastic anemia
- **How do differentiate between iron deficiency anemia & Minor thalassemia ?**
  - RDW index & Mentzer's index



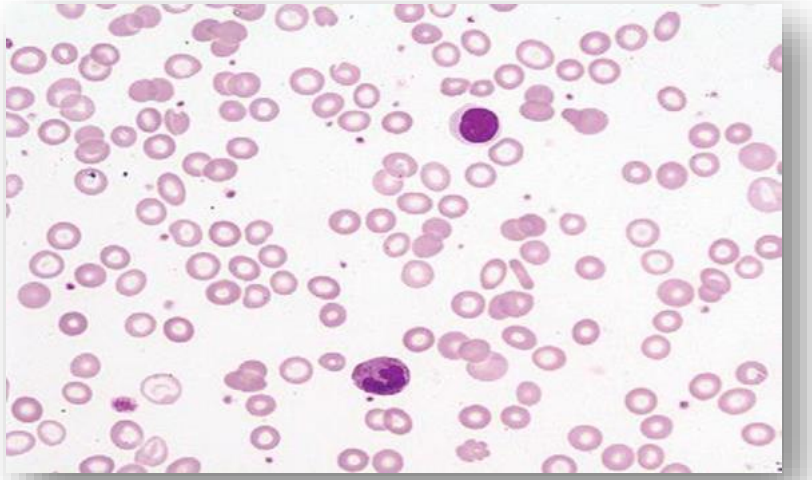
A blood film for an exclusively breast-fed baby

- **What is the type of this anemia ?**
  - Iron deficiency anemia
- **What other nutritional deficiencies might be seen in this patient ?**
  - Vitamin D
  - Fluoride



A blood film for a cow milk fed baby

- **What is the type of this anemia ?**
  - Iron deficiency anemia
- **Mention 2 tests to confirm your diagnosis**
  - Hemoglobin Electrophoresis, Ferritin Level

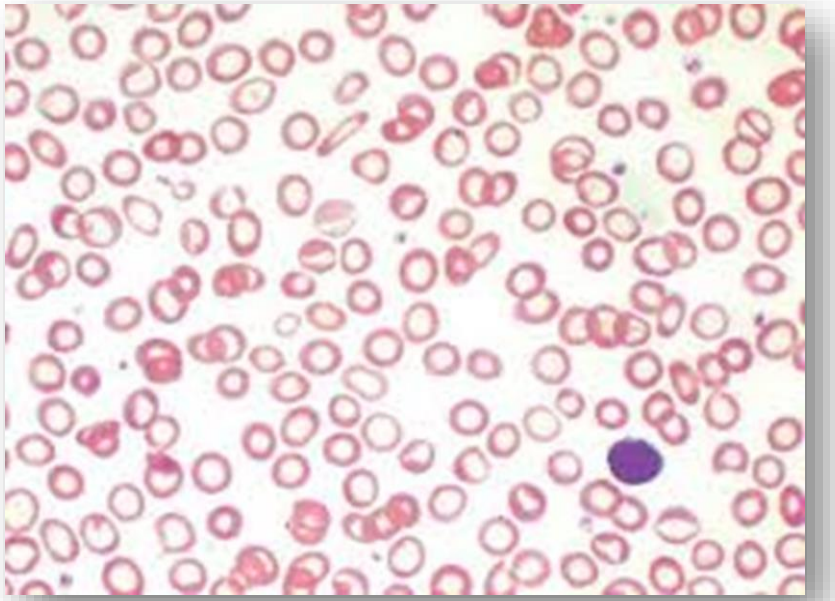


Iron deficiency anemia

- CBC of the 2-year-old male patient presented with pallor, Hb 6, RDW 19, MCV 55

- **What is the possible diagnosis ?**
  - Iron deficiency anemia

- **Mention helpful investigations to confirm the diagnosis**
  - Hemoglobin Electrophoresis, Ferritin Level

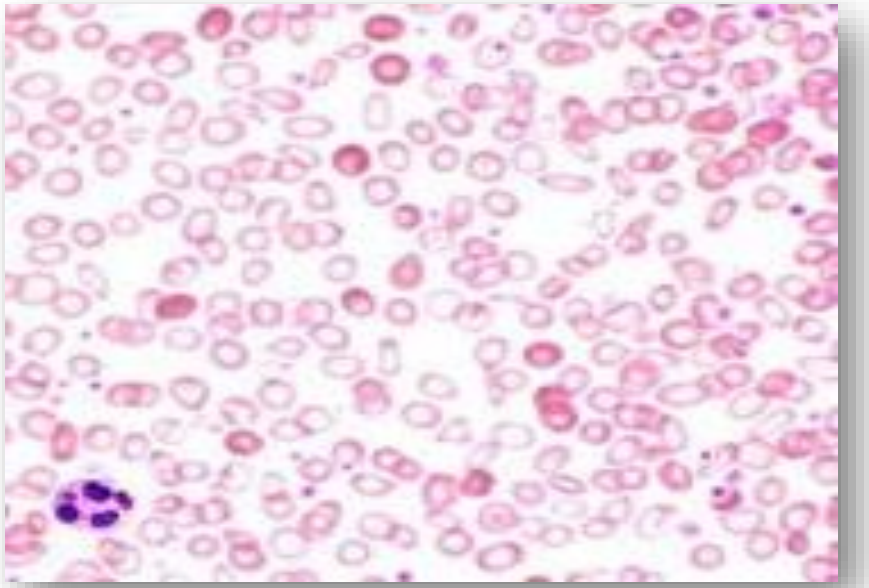


- **Reference normal values**
  - Hb >13.5,
  - RDW: 11–15%,
  - MCV: 80–100 μm^3

Thalassemia

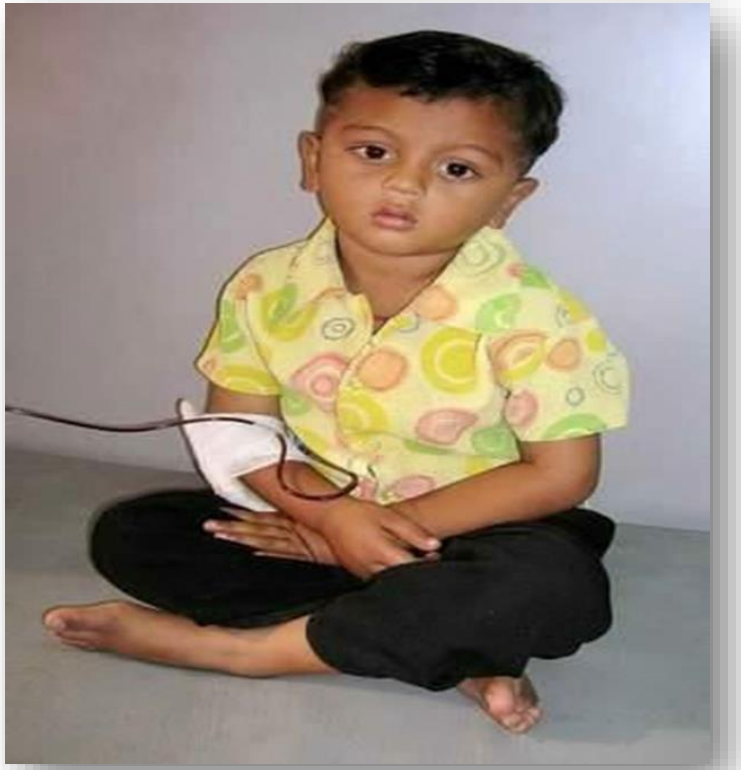
- Patient diagnosed with anemia and treated with iron supplements for 3 months then blood film performed

- **What is your diagnosis ?**
  - Thalassemia
- **How to confirm the diagnosis ?**
  - Hb electrophoresis



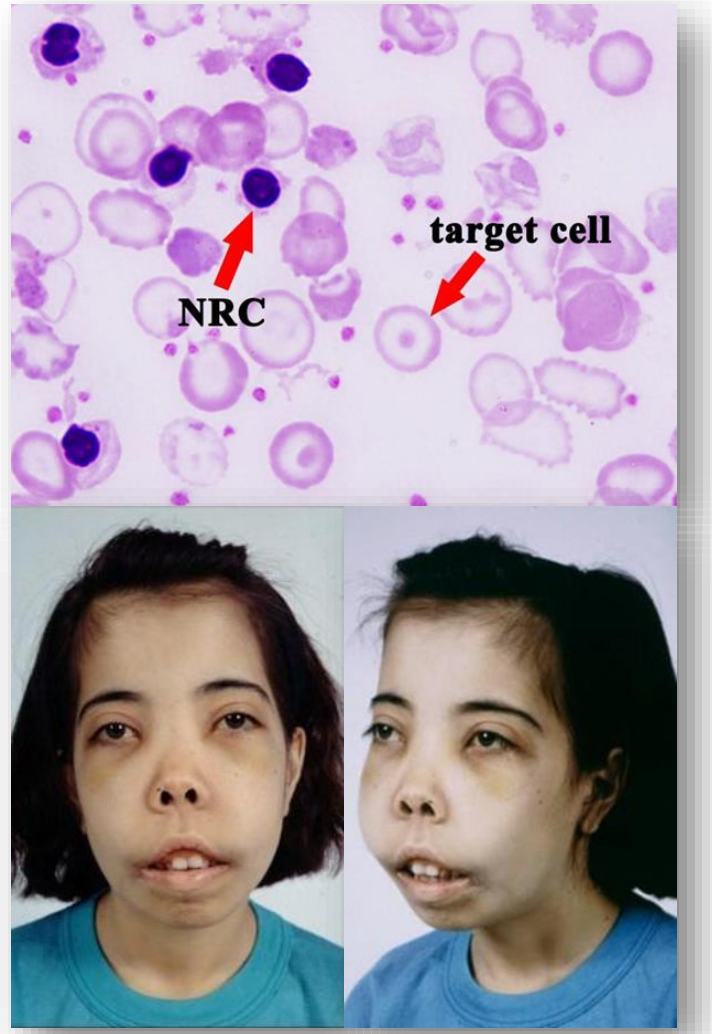
Anemic baby with splenomegaly

- **What is your diagnosis ?**
  - Thalassemia
  - Sickle cell anemia during splenic sequestration crisis
- **How to confirm the diagnosis ?**
  - Hb electrophoresis



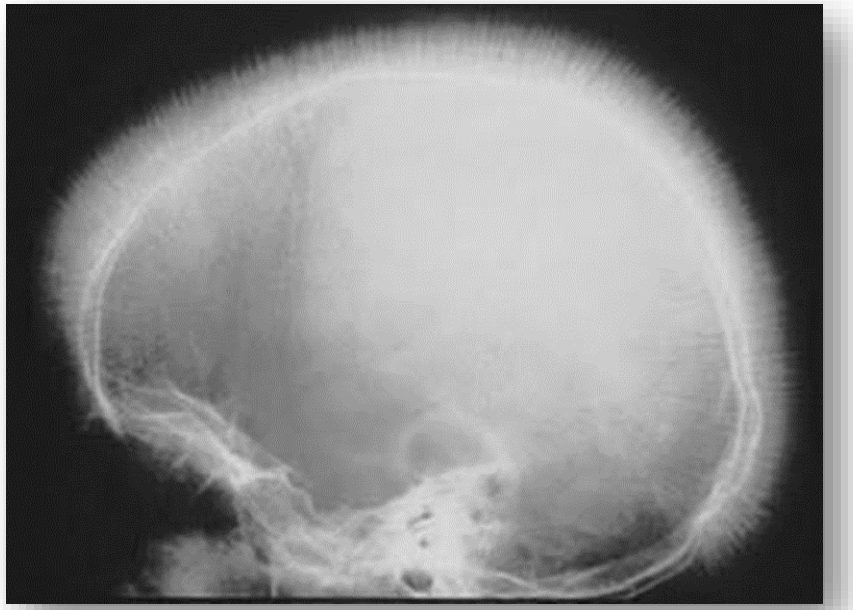
Thalassemia

- **What is the name of this face finding ?**
  - Chipmunk face
- **What is your diagnosis ?**
  - β-thalassemia major
- **Write two methods used in treatment ?**
  - Blood Transfusion, Bone marrow transplant



Thalassemia

- **What's this x-ray sign?**
  - Sun-ray or Hair-on-end or crew-cut appearance
- **What does it indicate?**
  - Expansion of hematopoiesis into the skull → β-thalassemia major, sickle cell anemia
- **What other 2 findings in the face you look for?**
  - Frontal bossing
  - Protruded maxilla



A 5-month-old boy is seen because of failure to thrive

- As part of the investigation the following blood tests are done

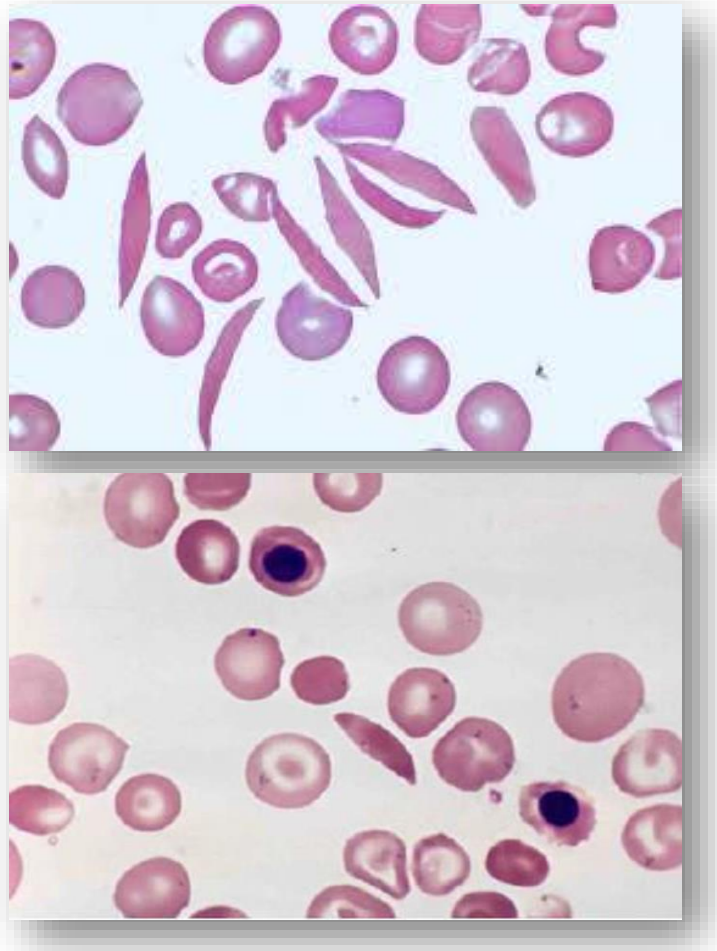
- **What is the diagnosis ?**
  - β-thalassemia major

- **What are the main treatment options?**
  - Regular blood transfusions with iron chelation therapy
  - Bone marrow transplantation

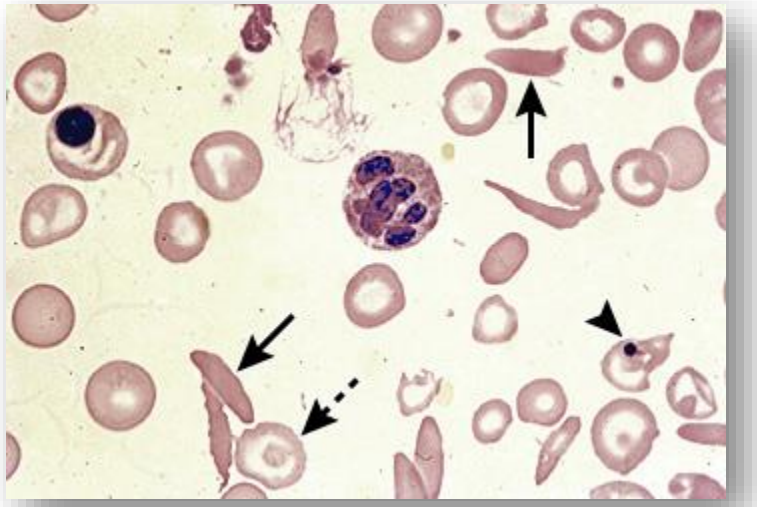
- Hb 4.2 g/dl
- WBC 12.3 x 109/l
- Plt 211 x 109/l
- Hemoglobin electrophoresis:
  - HbA - 0%
  - HbA2 - 9%
  - HbF - 91%

Sickle cell anemia

- **What does this blood film show ?**
  - Sickle cells
- **What is the type of inheritance ?**
  - Autosomal recessive
- **Mention a complication**
  - Vaso-occlusive crises
  - Hemolytic crises
  - Auto splenectomy



Sickle cell anemia



- 2-year-old male with pain, jaundice and pallor

❖Mention two crisis in this scenario

- Vaso-occlusive crisis and splenic sequestration crisis

❖Antibiotic in acute chest syndrome ?

- 3rd generation cephalosporin and macrolide

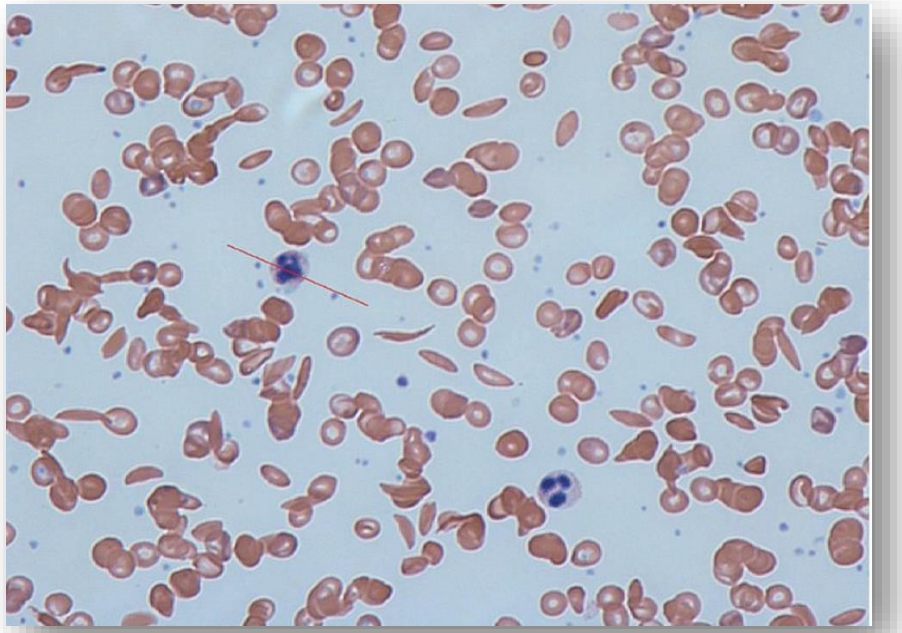
❖Mention 2 follow ups

- Immunizations
- Antibiotic prophylaxis against invasive pneumococcal disease until 5 years of age
- Hydroxyurea therapy regardless of clinical severity to minimize disease-related complications
- Annual transcranial doppler to screen for stroke risk from 2 months till 16 years of age
- Regular monitoring for other common complications of sickle cell disease

Sickle cell anemia

❖What's the next step in investigation?

- Hb Electrophoresis.



- ➢Sickle cell trait and/or disease is most commonly diagnosed on routine neonatal screening.
- ➢Confirmatory studies should be performed for all individuals with a positive or inconclusive result on initial screening.
- ➢Additional laboratory studies (CBC with MCV and reticulocyte count) and imaging are not required to confirm the diagnosis but may show characteristic changes.

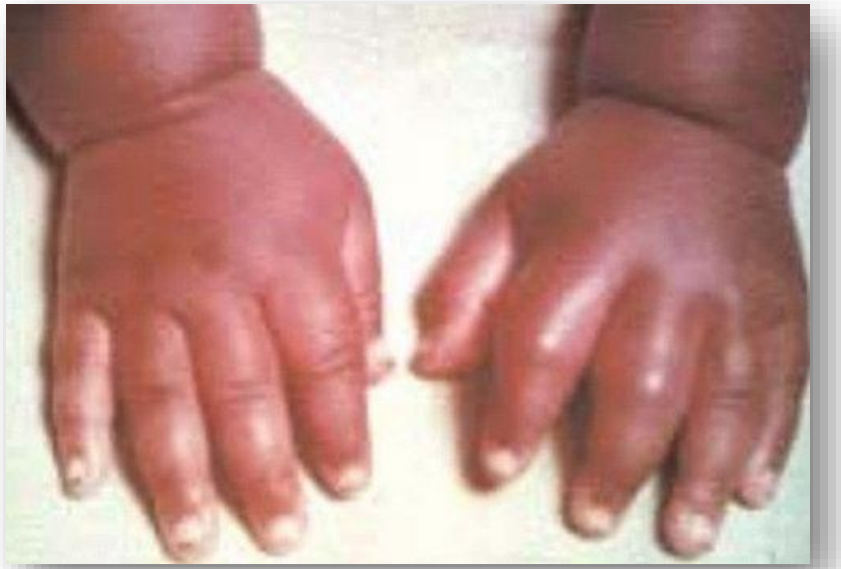
A 6-month-old child, presented with swollen fingers

❖**What is this sign ?**

- Dactylitis

❖**What is your spot diagnosis?**

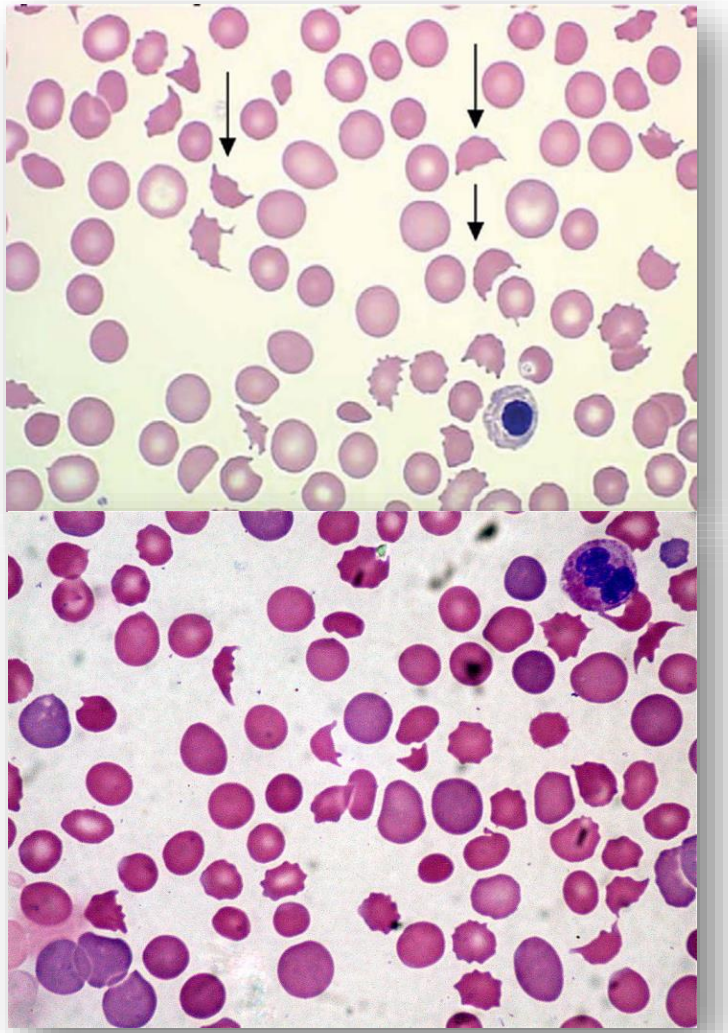
- Sickle cell anemia



➢Dactylitis is a common presenting sign of sickle cell anemia in infants and is due to small infarctions in the bones of the lower extremities

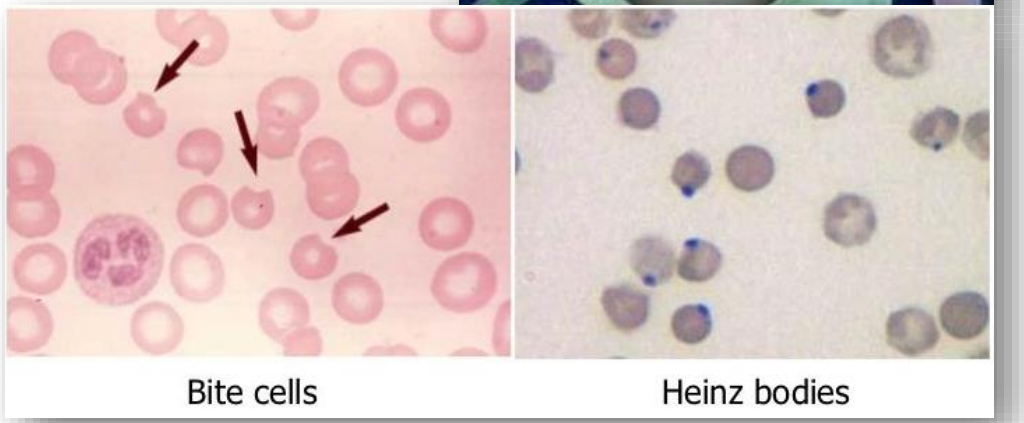
Schistocytes

- **What's the abnormality in this blood film ?**
  - Helmet cells (Schistocytes), Fragmented RBCs
- **Write 2 deferential diagnosis make this ?**
  - G6BD
  - HUS
  - DIC
  - any other hemolytic diseases



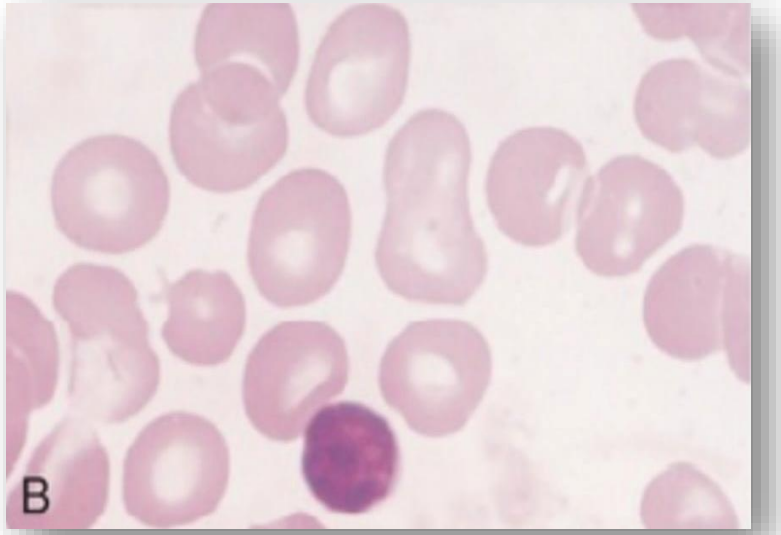
G6PD

- Scenario of child developed jaundice after treated with nitrofurantoin for his UTI, he was pale with low hemoglobin, with Blood film
- **Findings on Blood film ?**
  - Bite cell, Heinz bodies
- **Single best test to confirm your diagnosis ?**
  - G6PD enzyme level
- **What is the management ?**
  - Avoid triggers
  - Blood transfusions only in severe cases



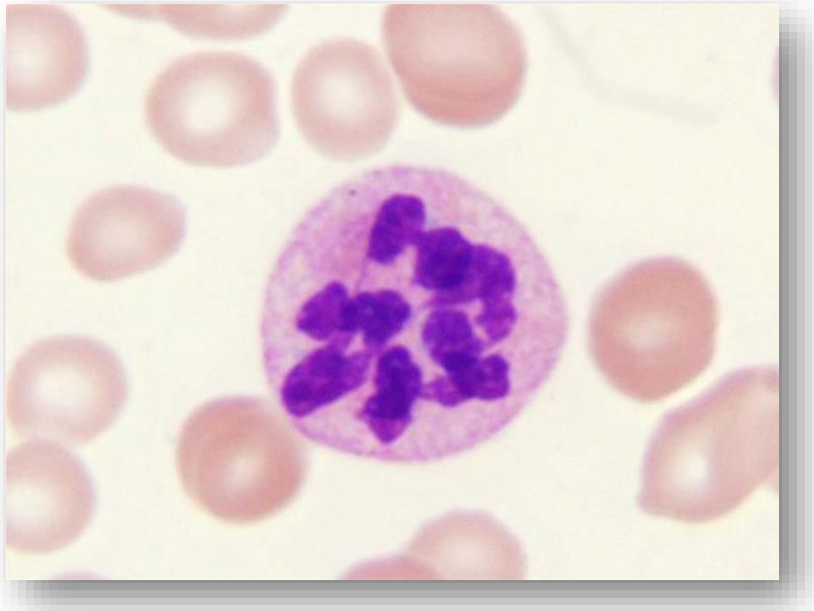
Macrocytic anemia

- **Describe what you see ?**
  - Macrocytic cell
- **Write 2 differential diagnoses**
  - Folic acid deficiency
  - Vitamin B12 deficiency



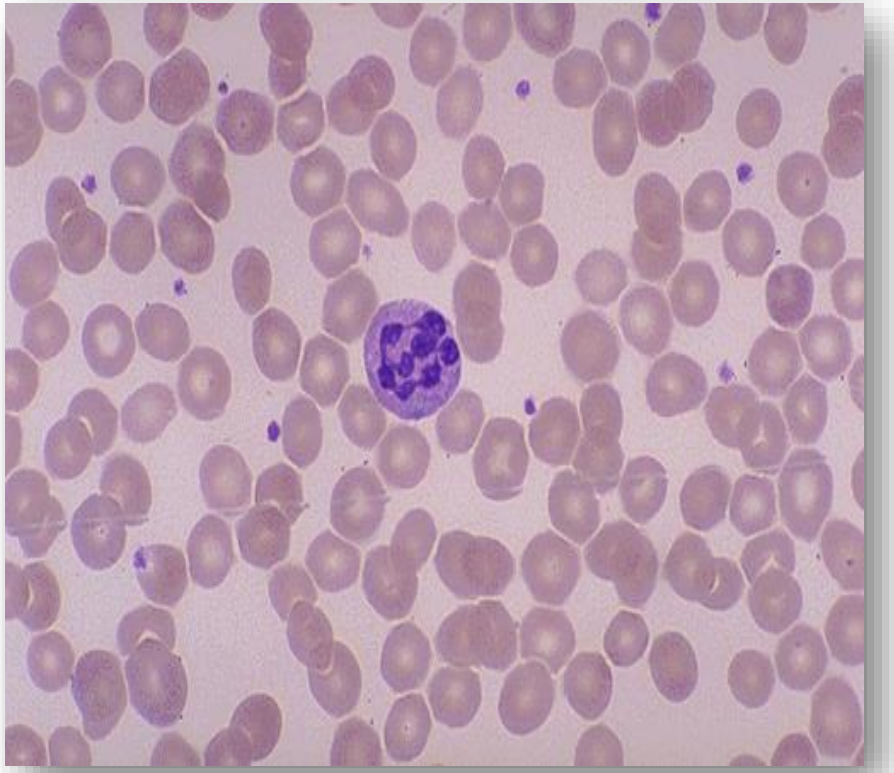
Megaloblastic macrocytic anemia

- **Describe what you see in the picture.**
  - Hyper-segmented Neutrophil.
- **What's this condition?**
  - Megaloblastic macrocytic anemia
- **What is the deficient nutrient ?**
  - B_{12} or Folate



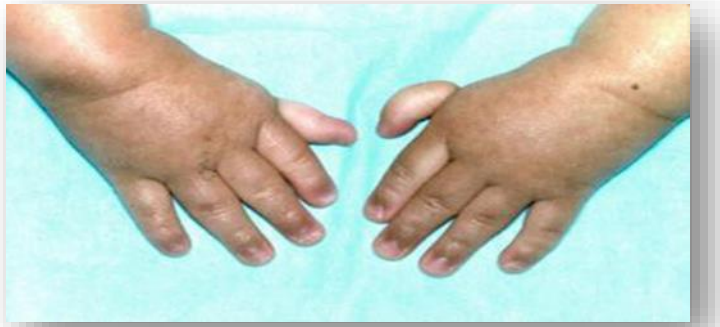
Megaloblastic macrocytic anemia

- **Which type of anemia ?**
  - Megaloblastic macrocytic anemia
- **Mention a risk factor ?**
  - B12 deficiency, folate deficiency
- **lab test need for conformation**
  - Vitamin B12 and folate levels
  - If normal labs suspect liver disease



Fanconi anemia

- **What is your diagnosis ?**
  - Fanconi anemia
- **What other abnormalities you may find in this patient ?**
  - Short stature
  - Abnormal thumb
  - Renal & cardiac anomalies
- **What's the definitive diagnostic test ?**
  - White cell chromosomal fragility, high HbF



Fanconi anemia

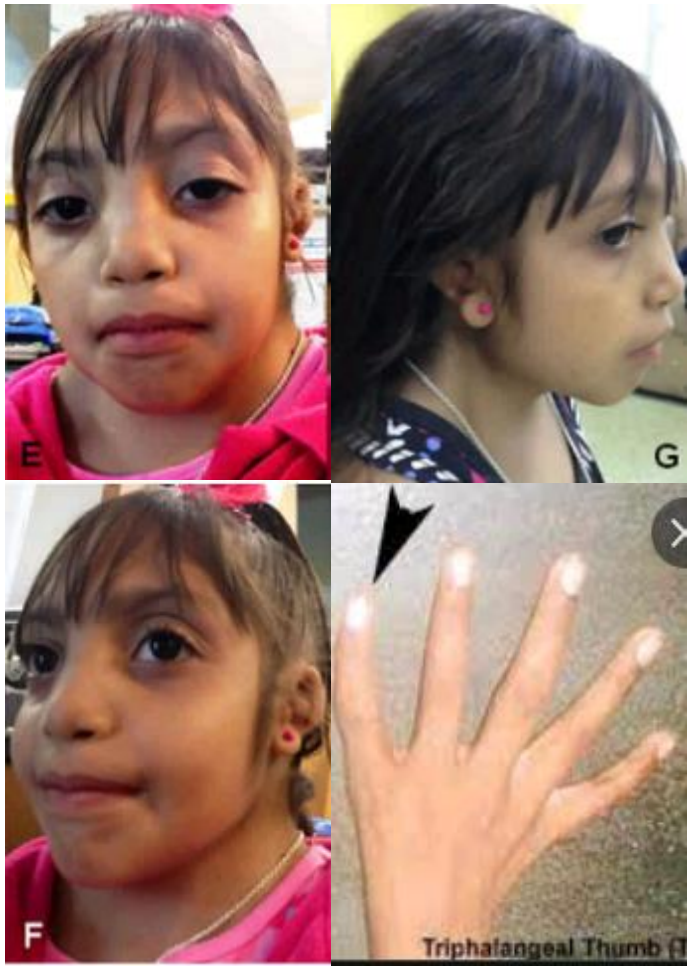
- **Describe the abnormality in the picture**
  - Absent thumb
- **Name the condition associated with this abnormality**
  - Fanconi's anemia

Fanconi anemia

- Hereditary autosomal recessive disorder due to a DNA crosslink repair defect resulting in bone marrow failure
- Skeletal and organ abnormalities: short stature, hypo- and hyperpigmentation, cafe-au-lait spots, microcephaly, developmental delay, thumb and forearm malformations, kidney, GI, heart, eye, and ear abnormalities
- Laboratory tests show pancytopenia and normocytic or macrocytic anemia.
- ~ 50% of patients with Fanconi anemia will develop acute myeloid leukemia or myelodysplastic syndromes in early adulthood.

Diamond-Blackfan Anemia

- Intrinsic defect of erythroid progenitor cells → ↑ apoptosis (Pure red cell aplasia)
- Usually, autosomal dominant inheritance
- Rapid onset of macrocytic (nonmegaloblastic) anemia in infancy (usually diagnosed within the 1st year of life)
- **Additional clinical features**:
  - Short stature, webbed neck
  - Upper extremity malformations (e.g., triphalangeal thumbs)
  - Microcephaly, micrognathia
  - Hypertelorism, flat nasal bridge, cleft palate
  - Congenital cataracts or glaucoma
  - Atrial and ventricular septal defects



Bleeding disorders

Low-Yield

Platelets disorders

- **Immune thrombocytopenic purpura (ITP)**
- **Microangiopathic hemolytic anemia**
  - Thrombotic thrombocytopenic purpura (TTP)
  - Hemolytic uremic syndrome (HUS)
- **Qualitative platelet disorders**
  - Bernard-Soulier syndrome (GpIb deficiency)
  - Glanzmann thrombasthenia (GpIIb/IIIa deficiency)
  - Uremic platelet dysfunction
- **Quantitative platelet disorders**
  - **W**iskott-**A**ldrich syndrome: triad of **T**hrombocytopenia, **E**czema, **R**ecurrent pyogenic infections
  - Disseminated intravascular coagulation
  - Thrombocytopenia with absent radii syndrome
  - Congenital amegakaryocytic thrombocytopenia
  - Bone marrow failure
  - leukocyte adhesion deficiency

Coagulation disorders

- **von Willebrand disease** is the most common congenital bleeding disorder.
- **Hemophilias**
  - Hemophilia A: Factor 8, X-linked recessive
  - Hemophilia B: Factor 9, X-linked recessive
  - Hemophilia C: Factor 11, autosomal recessive
  - Factor VIII and factor IX deficiencies (Hemophilias) are the most common severe inherited bleeding disorders.
- **Factor XIII Deficiency**
  - Factor XIII is the longest half-life of all the clotting factors (10 days).
  - Screening coagulation tests (PT and PTT) are normal.
  - Factor XIII deficiency is associated with poor wound healing.
  - Delayed separation of umbilical stump (can also occur with leukocyte adhesion deficiency) and intracranial hemorrhage

Immune Thrombocytopenic Purpura

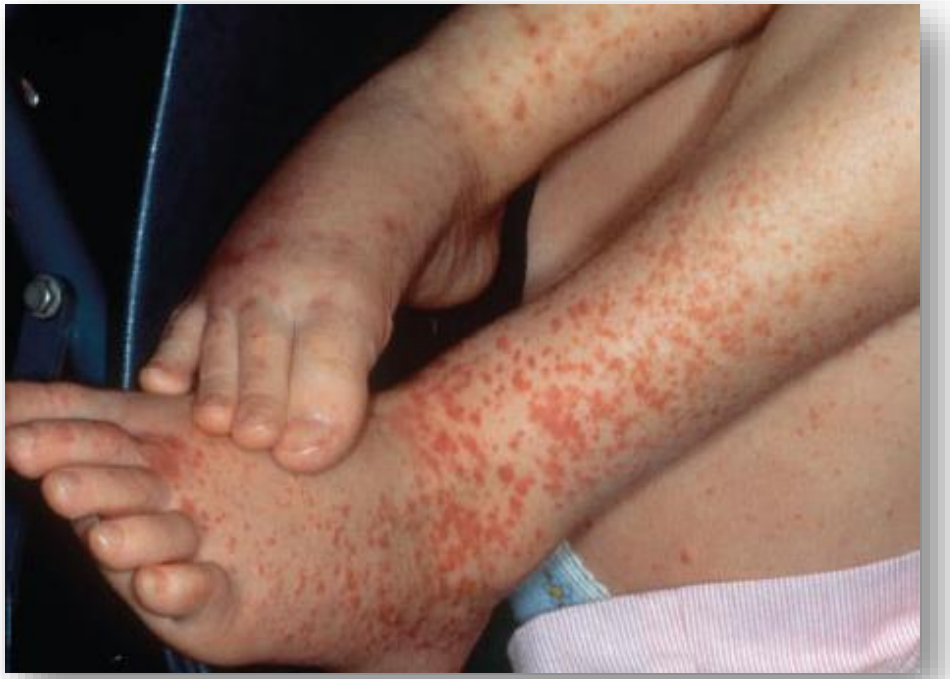
- The most common cause of thrombocytopenia in children and adults
- **Etiology**: commonly follows viral infection (acute form)
- **Pathophysiology**:
  - It is due to autoimmune production of IgG (by plasma cells in the spleen) against GPIIb/IIIa
  - The antibody-bound platelets are consumed by splenic macrophages, resulting in thrombocytopenia
- **Subtypes**:
  - The **acute** form of ITP commonly arises in children, weeks after a viral infection or immunization
  - The **chronic** form of ITP commonly arises in women of childbearing age, and it may be primary or secondary (e.g., SLE)

Immune Thrombocytopenic Purpura

- **Lab findings**:
  - Increased bleeding time / PFA; due to decreased platelet count
  - Normal PT and normal PTT
- **Bone marrow biopsy**: increased megakaryocytes
- **Management**:
  - **Observation**: Children with no symptoms or only minor mucocutaneous bleeding
  - **Corticosteroids**: Patients with significant non-life-threatening mucosal bleeding or symptoms impacting quality-of-life
  - **Corticosteroids + IVIG + Platelets transfusion as needed**: Patients requiring emergency treatment (Life-threatening bleeding, acute neurological features or anticipated urgent surgery or invasive procedure)

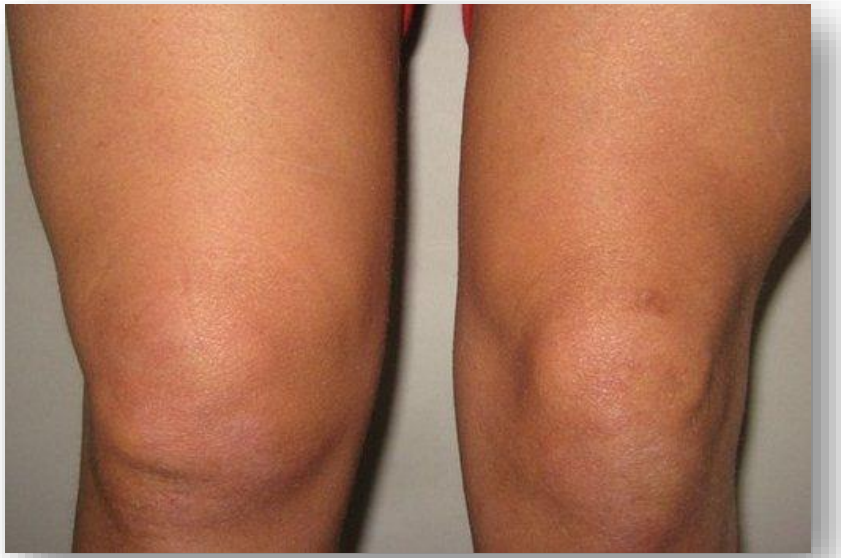
Immune Thrombocytopenic Purpura

- History of skin rash in 12 years old girl for 2 days. Her platelet count 4000
- **Your differential diagnosis**
  - ITP
- **Your treatment**
  - Observation



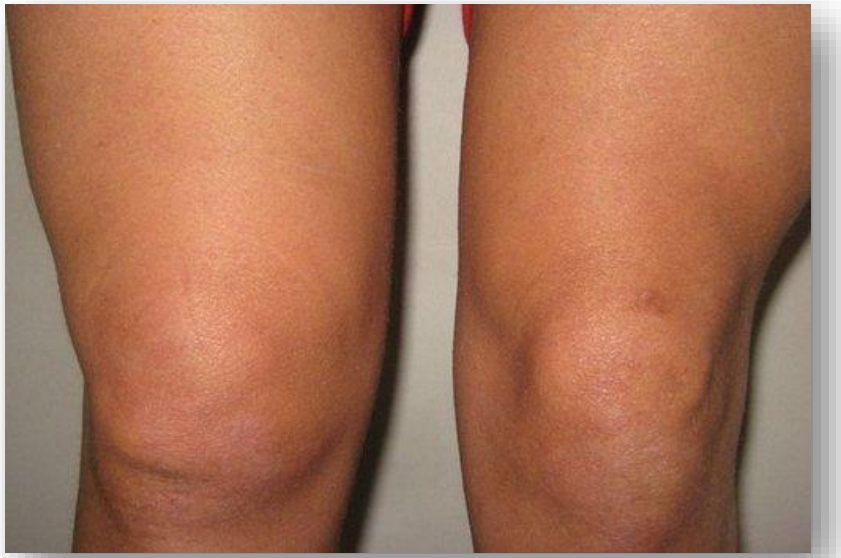
Hemophilia

- **Write one differential diagnosis**
  - Hemophilia
- **What u expect to see in the coagulation profile for this patient ?**
  - Prolonged PTT
  - Normal PT
  - Normal bleeding time
- **Treatment**: Give deficient factors
- **Note**: Hemophilia A & B are X-linked recessive, whereas Hemophilia C is autosomal recessive



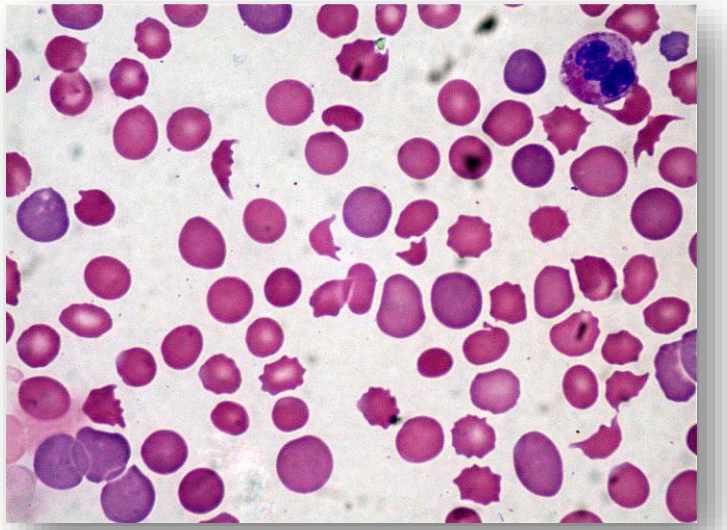
Knee joint

- **Mention 3 DDx**
  - Trauma, septic arthritis, hemophilia, oligoarticular JIA, HSP, …
- **What Lab test you'd like to order ?**
  - CBC, ANA, coagulation profile, …
- **Mention 3 physical sign**
  - Tenderness, limitation of movement, Rash, …



1-year-old male, acute illness, severe sepsis

- **What's the abnormality in this blood film ?**
  - Helmet cells (Schistocytes), Fragmented RBCs
- **Lab findings suggest the picture**
  - Schistocytes
  - Increased fibrin degradation products (D-dimers)
  - Decreased fibrinogen
  - Decreased factor V & VIII (in addition to other factors)
    - These are specific findings, other findings such as increased PTT PT/INR are also present
- **What is your diagnosis ?**
  - DIC

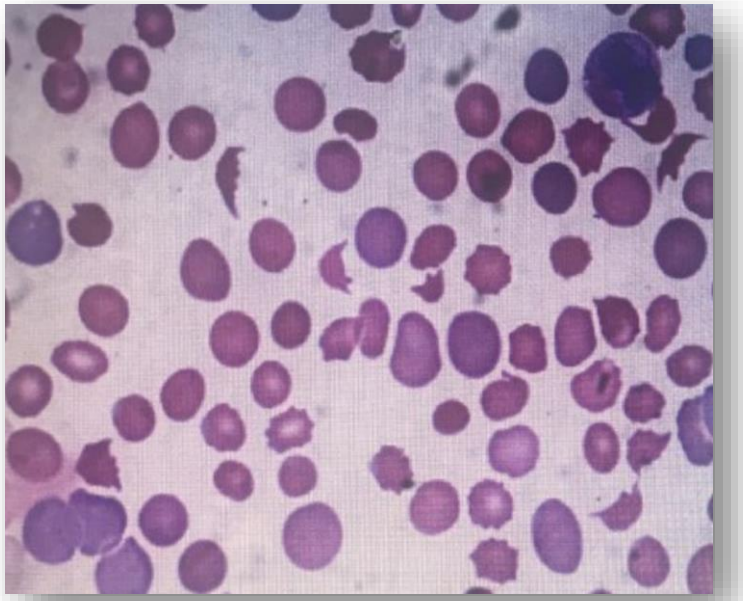


SSSTOP Making New Thrombi

- **S**: **S**troke, **S**nake bites, **S**epsis
- **T**: **T**rauma
- **O**: **O**bstetric complications
- **P**: **P**ancreatitis
- **M**: **M**alignancy
- **N**: **N**ephrotic
- **T**: **T**ransfusion

DIC

- 3 years old boy is admitted to the PICU, He looks ill and pale, BP is 145 / 96, pulse 148,RR 38 AND pulse ox 95% off Oxygen, he has history of gastroenteritis few days ago. Lab results: Na 132, K 3.4, urea 104 mg/dl, creatinine 2.8 mg/dl, glucose 87 mg / DL, alt 28 U/L, albumin 3.7, blood film is shown.



- **Which of the following is not consistent with the diagnosis ?**

a. Elevated lactate dehydrogenase LDH
b. Increased fibrinogen degradation products FDP
c. Normal C3 level
d. Presence of hematuria
e. Thrombocytopenia

Hematological malignancies

Low-Yield

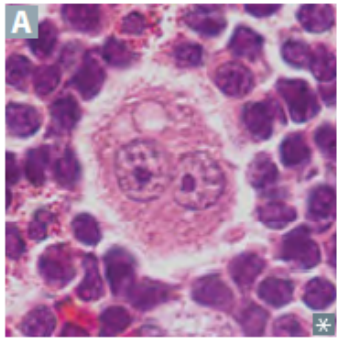
إضافي

Hodgkin vs non- Hodgkin lymphoma

Hodgkin vs non-Hodgkin lymphoma

Hodgkin	Non-Hodgkin
Both may have constitutional ("B") signs/symptoms: low-grade fever, night sweats, weight loss.	
Localized, single group of nodes with contiguous spread (stage is strongest predictor of prognosis). Better prognosis.	Multiple lymph nodes involved; extranodal involvement common; noncontiguous spread. Worse prognosis.
Characterized by Reed-Sternberg cells.	Majority involve B cells; rarely of T-cell lineage.
Bimodal distribution: young adults, > 55 years.	Can occur in children and adults.
Associated with EBV.	May be associated with autoimmune diseases and viral infections (eg, HIV, EBV, HTLV).

Hodgkin lymphoma

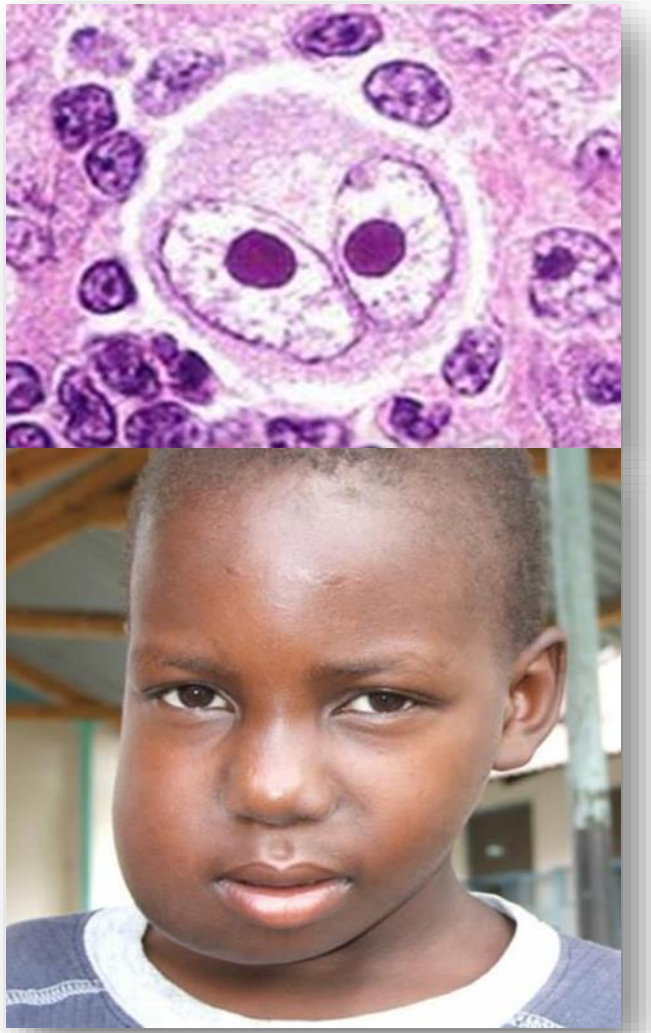


Contains Reed-Sternberg cells: distinctive tumor giant cells; bilobed nucleus with the 2 halves as mirror images ("owl eyes" A). RS cells are CD15+ and CD30+ B-cell origin. 2 owl eyes × 15 = 30.

SUBTYPE	NOTES
Nodular sclerosis	Most common
Mixed cellularity	Eosinophilia; seen in immunocompromised patients
Lymphocyte rich	Best prognosis (the rich have better bank accounts)
Lymphocyte depleted	Worst prognosis (the poor have worse bank accounts); seen in immunocompromised patients

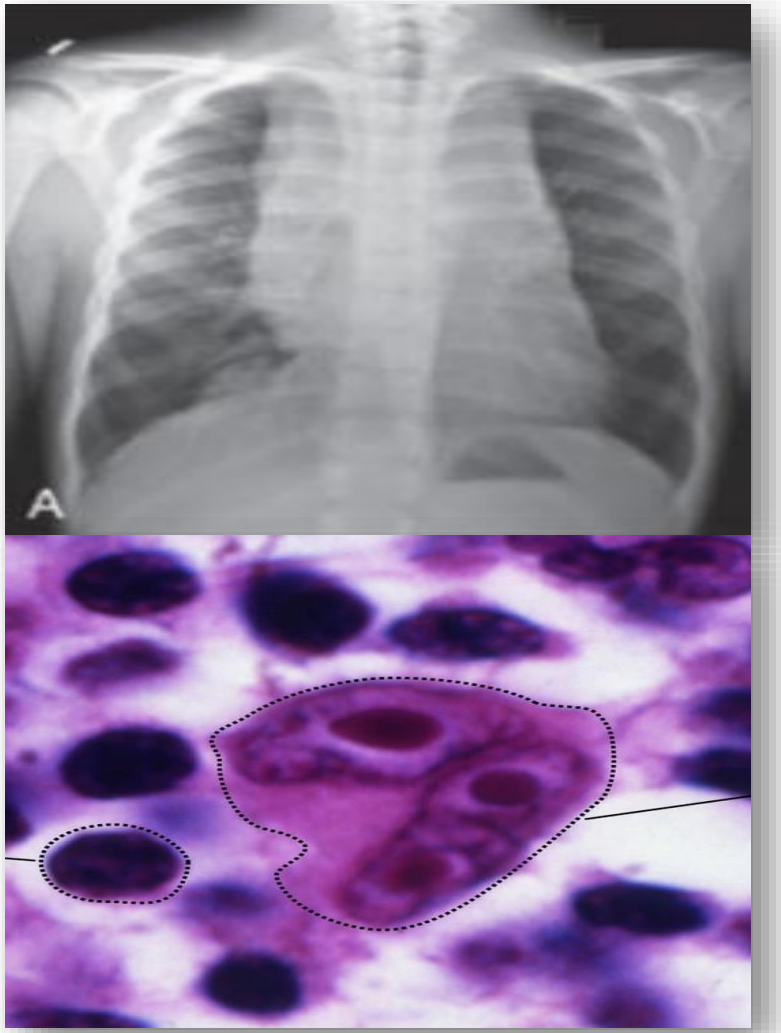
Hodgkin's lymphoma

- **What is the name of the cell on the biopsy?**
  - Reed Sternberg cell
- **What is your diagnosis?**
  - Hodgkin's lymphoma
- **What is the mainstay of treatment ?**
  - Radiation



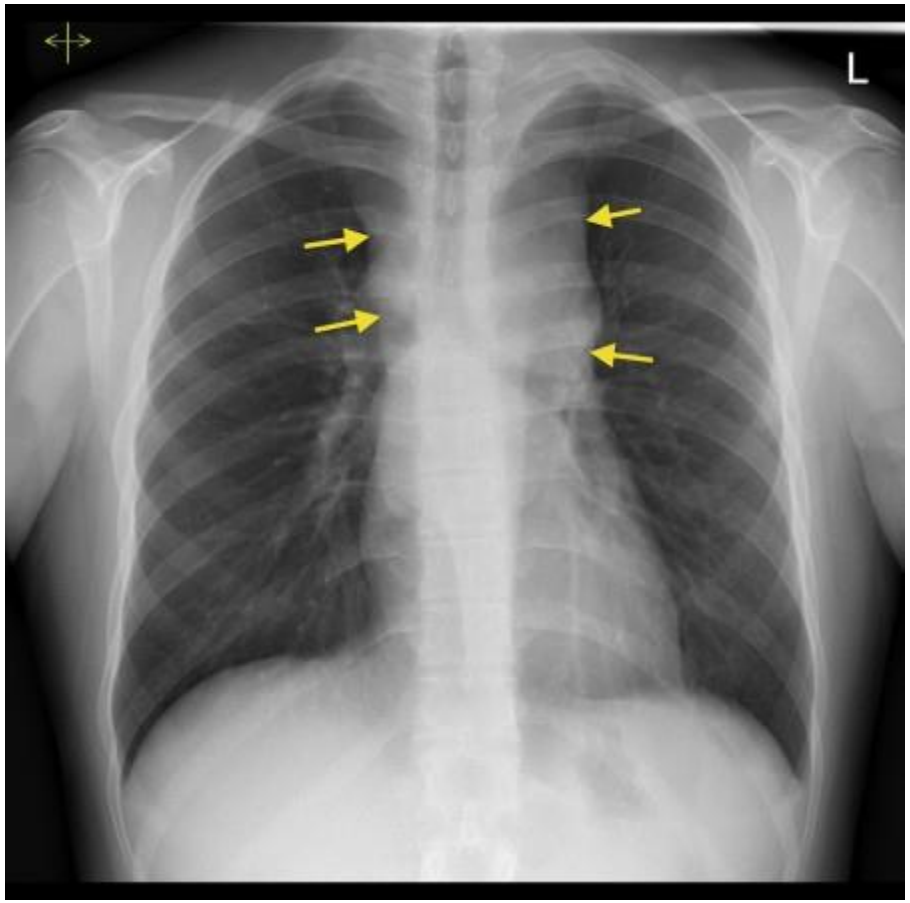
Hodgkin's lymphoma

- child complains of cough, on examination there was lymphadenopathy
- **Describe what you see X-ray**
  - Wide mediastinum
- **What is the cell shown ?**
  - Reed Sternberg cell
- **What is your Dx ?**
  - Hodgkin's lymphoma
- The nodular sclerosis subtype of Hodgkin lymphoma typically presents as an enlarging cervical or mediastinal lymph node in a female



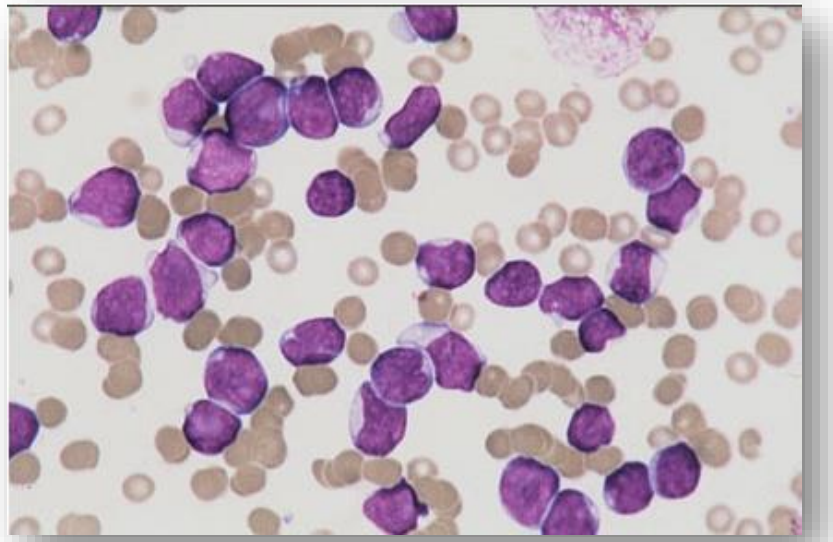
Hodgkin's lymphoma

- **What's the finding in this CXR ?**
  - Widening of the mediastinum
- **Mention 2 malignant causes**
  - Lymphoma
  - Germ cell tumor (Teratoma)
  - Thymoma



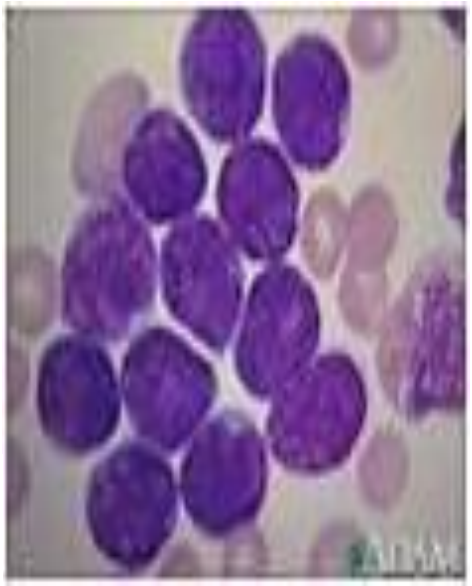
Acute Lymphocytic Leukemia

- **Mention 2 Acute complications of this disease?**
  - Bleeding
  - Acute tumor lysis syndrome
  - Thrombosis
  - Serious infection

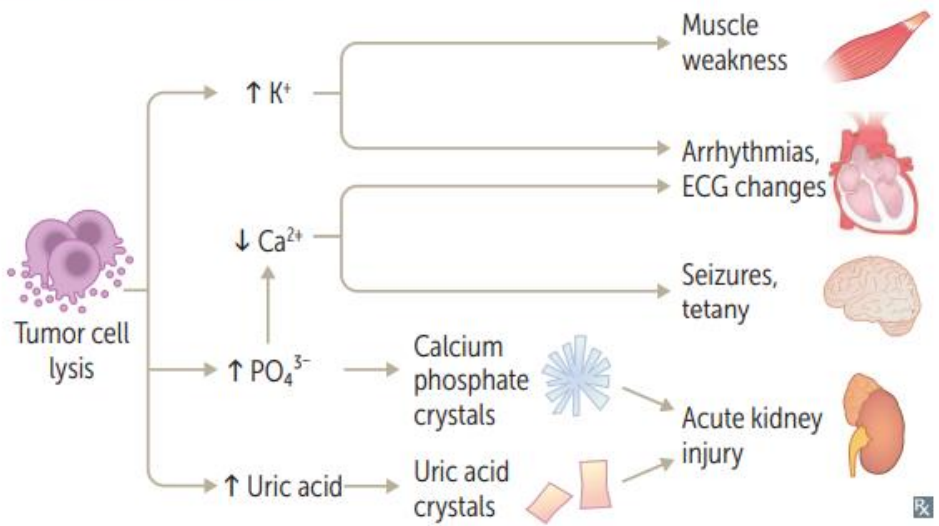


Child present with low grade fever and limping

- **Give two other presentation**
  - Purpura, lymphadenopathy, bone pain, pallor
- **3 electrolyte affected in tumor lysis syndrome**
  - Elevated uric acid, K, PO4, and decrease Ca



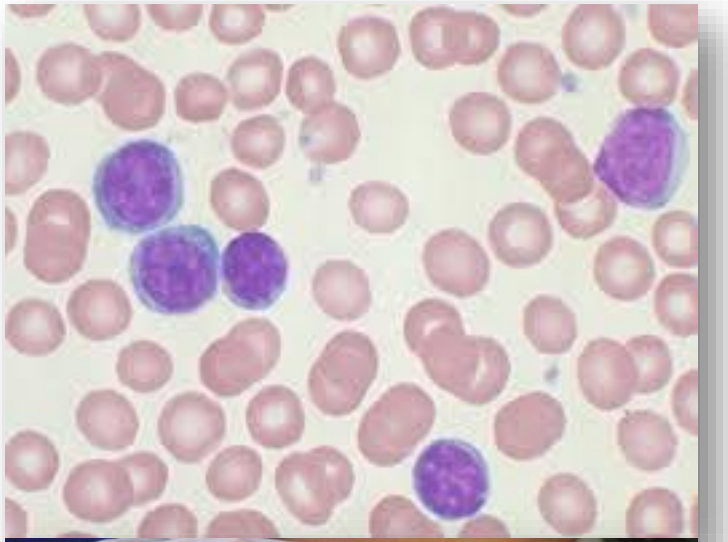
Tumor lysis syndrome

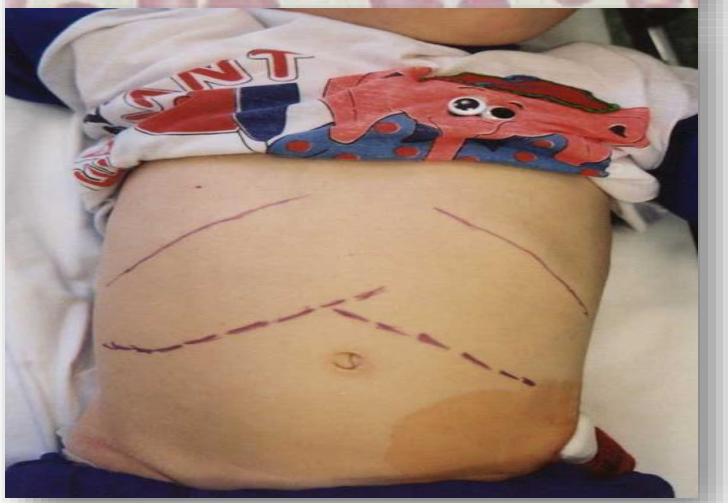


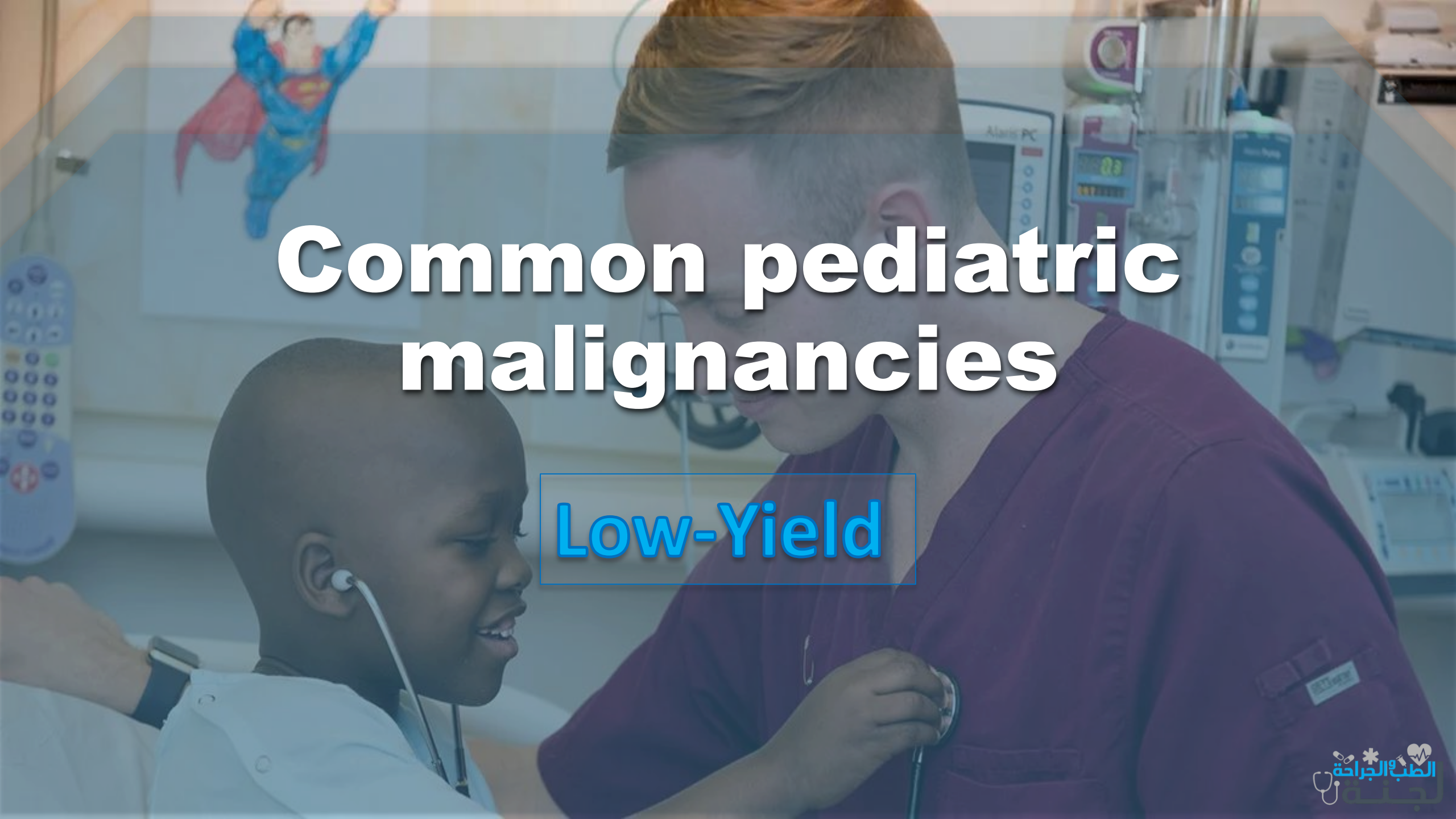
Oncologic emergency triggered by massive tumor cell lysis, seen most often with lymphomas/leukemias. Usually caused by treatment initiation, but can occur spontaneously with fast-growing cancers. Release of K^+ → hyperkalemia, release of PO_4^{3-} → hyperphosphatemia, hypocalcemia due to Ca^{2+} sequestration by PO_4^{3-}. ↑ nucleic acid breakdown → hyperuricemia → acute kidney injury. Prevention and treatment include aggressive hydration, allopurinol, rasburicase.

Acute Lymphocytic Leukemia

- **What is the type of cells pointed at?**
  - Lymphoblasts
- **Found in which type of malignancy?**
  - ALL
- **Mention 3 good prognostic factors for this disease?**
  - Age: 1-10 year.
  - WBC: < 50,000
  - Chrom. Abnormalities: presence of TEL/AML1 gene. Philadelphia- negative, hyperdiploidy, absence of MLL rearrangement
  - Immunophenotype: B-cell ALL
  - No CNS involvement
  - Early Response to Chemotherapy





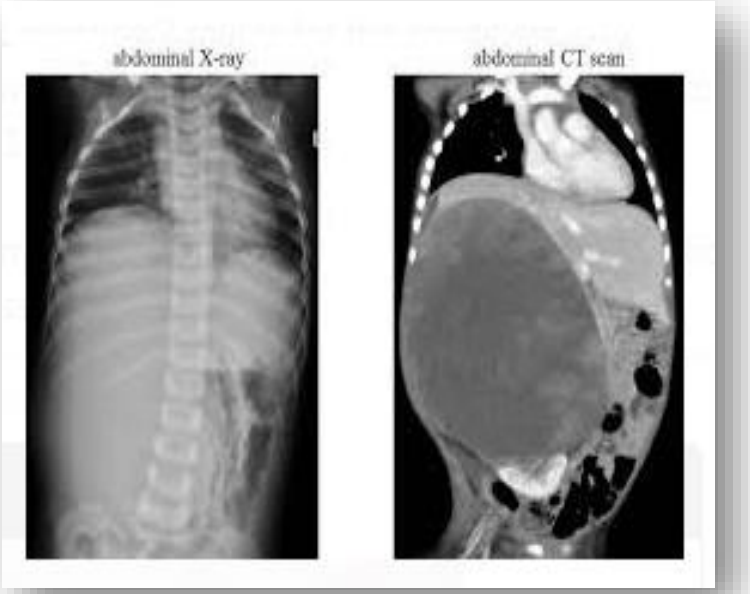


Common pediatric malignancies

Low-Yield

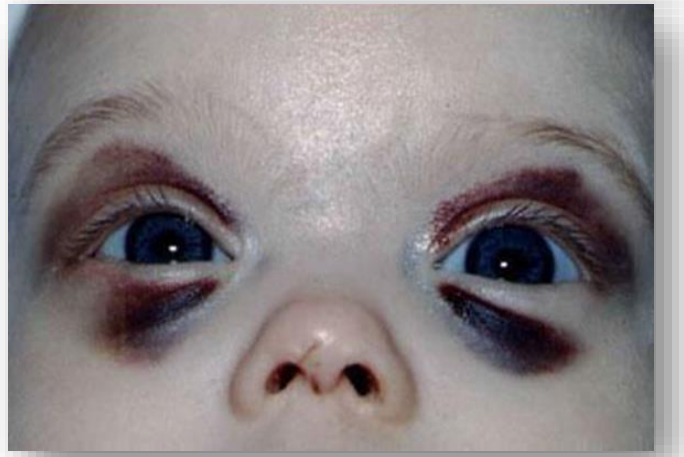
Patient present with hematuria & flank pain

- **Mention two DDx:**
  - Neuroblastoma
  - Wilms tumor
- **Mention 2 other clinical manifestations**
  - Displacing mass, mainly confined to the flank
- **Mention associated syndrome**
  - Blackwith-Wiedmann syndrome
  - WAGR syndrome
- **What can you see in his eyes ?**
  - Aniridia



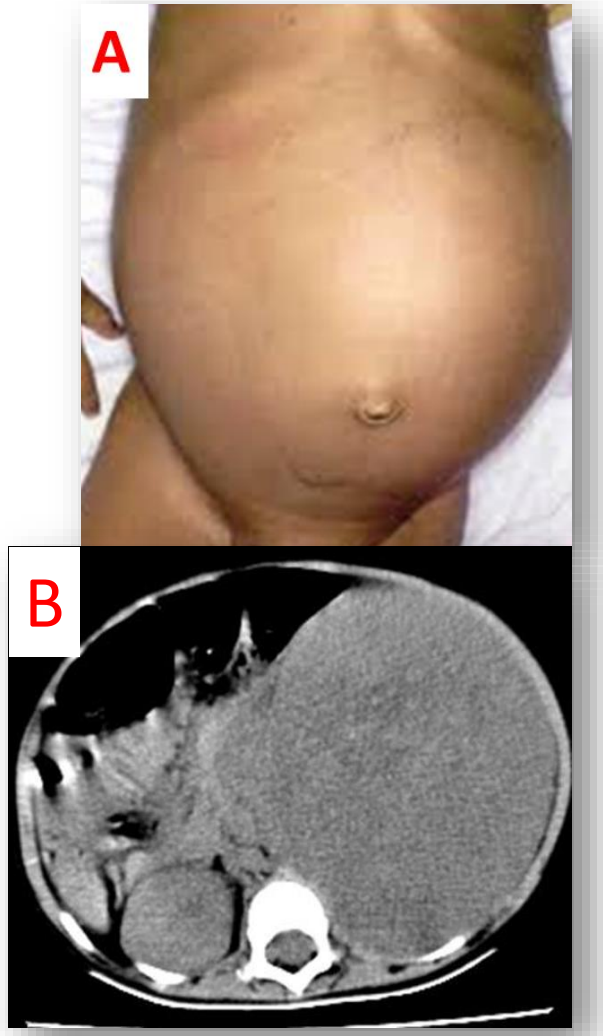
Hx of abdominal pain

- **What other systems would you like to examine?**
  - CNS, Respiratory or GI.
- **What's the most likely Dx. ?**
  - Neuroblastoma.
- **Mention 2 non-radiological investigations**
  - VMA, Bone marrow biopsy.



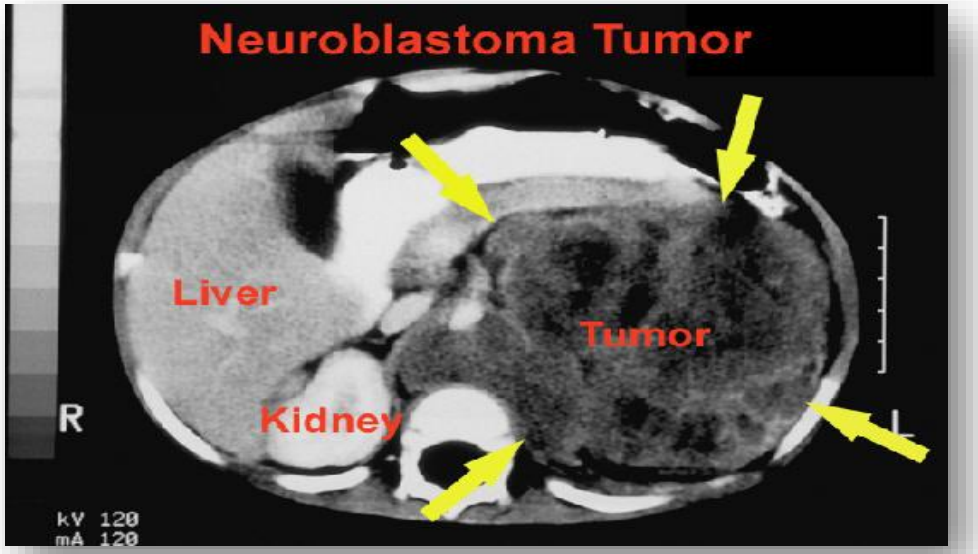
Wilms tumor

- **Describe what you see in A**
  - Asymmetrical abdomen
- **Describe what you see in B**
  - Abdominal CT-scan with large left intra-renal mass
- **What is your diagnosis ?**
  - Nephroblastoma (Wilms tumor)
- **What is the stage 5 of this disease ?**
  - Bilateral renal involvement



CT scan for a 3-year-old male boy presented with unilateral kidney mass

- **Describe the mass**
  - Left abdominal mass, heterogeneous, crossing midline, irregular shape
- **What is the most likely diagnosis ?**
  - Neuroblastoma



Neuroblastoma Vs Wilms Tumor

	Wilms Tumor	Neuroblastoma
Location	Abdominal mass	Abdominal mass
Origin	Intrarenal	Extrarenal
Physical examination	Displacing mass, mainly confined to the flank	Non-mobile mass, more likely to cross the midline
Pattern of spread	Direct expansion with displacement of adjacent structures	Encasement of vessels and aortic elevation
Other	• Intrinsically displaces urinary collecting systems • Asymptomatic • Macroglossia • Hypertension & hematuria	• Externally displaces kidney • Neural extension • Often calcified (90%) • Irritable child, tender • Raccoon eyes

إضافي

Wilms Tumor Staging

National Wilms Tumor Study (NWTS) system [8]			
Stage	Tumor location	Tumor spread	Surgery
I	• Unilateral	• Limited to the kidney • No renal sinus vessel involvement	• Complete resection with no tumor beyond the excision margins

II	• Unilateral	• Extends beyond kidney
		  ◦ Renal capsule
		  ◦ Soft tissue of the renal sinus
		  ◦ Blood vessels beyond the renal parenchyma	

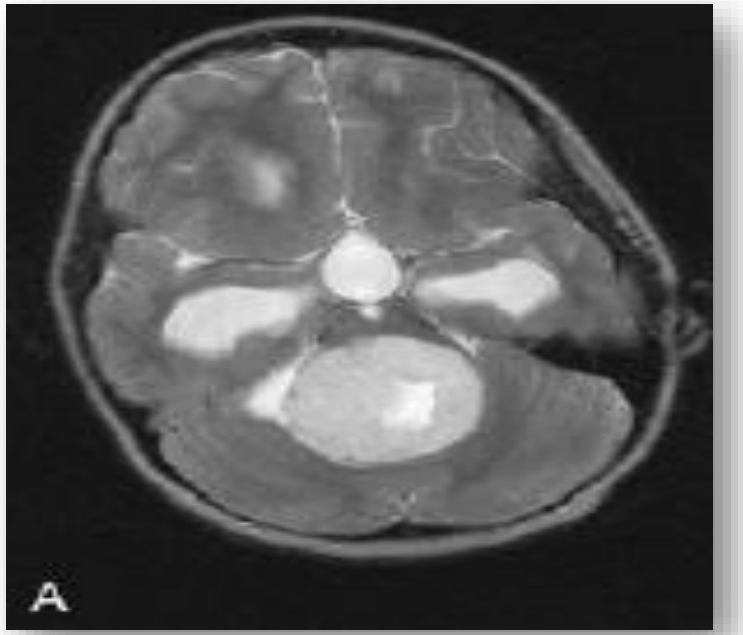
III	• Unilateral	• Confined to the abdomen • Dissemination in abdomen (e.g., regional lymph nodes, peritoneal involvement)	• Incomplete resection with residual tumor remaining postoperatively (e.g., not resectable due to infiltration into vital structures)
IV	• Unilateral	• Hematogenous metastasis (e.g., lung, liver, bone, brain) • Distant lymph nodes involvement (outside of abdomen)	
V	• Bilateral	• Both kidneys are affected	

Neuroblastoma Staging

International Neuroblastoma Staging System (INSS) [13]	
Stage	**Definition**
1	• Localized tumor • Complete gross excision with or without microscopic residuals • Negative ipsilateral lymph nodes
2A	• Localized tumor • Incomplete gross excision • Negative ipsilateral lymph nodes
2B	• Localized tumor • Complete or incomplete gross excision • Positive ipsilateral lymph nodes
3	• Unresectable unilateral tumor that crosses the midline with or without lymph node involvement • Any tumor with positive contralateral lymph nodes • Midline tumor with bilateral tumor or lymph node involvement
4	• Any tumor with dissemination to distant lymph nodes or other organs (e.g., bone, liver, skin), with the exception of Stage 4S disease
4S	• Localized primary tumor with dissemination to skin, liver, or bone marrow, occurring in infants < 12 months

Brain tumor

- **What is the abnormality in this picture?**
  - Brain tumor
- **The patient comes complaining from what ?**
  - Headache, irritability, lethargy.
  - Loss of vision
  - Ataxia, posture.



Infectious diseases

Exanthem

صورة يعرفها كل من درس محاضرات دكتورة غادة

High-Yield

Exanthem

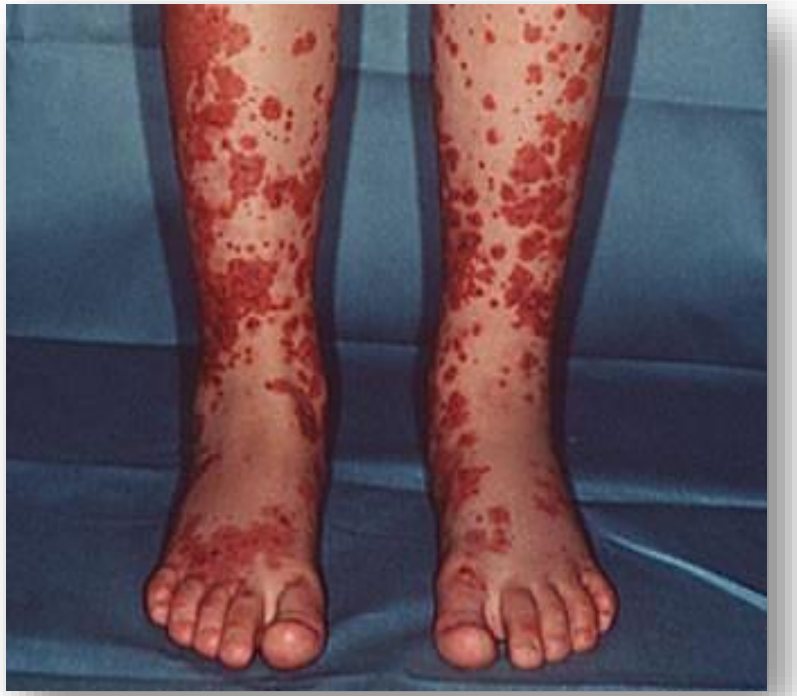
- An exanthem is a widespread rash occurring on the outside of the body and usually occurring in children.
- An exanthem can be caused by toxins, drugs, or microorganisms, or can result from autoimmune disease.
- We will talk about:
  - Diseases with rashes: Henoch-Schönlein Purpura, Kawasaki, & ITP
  - Infectious exanthem

Henoch-Schönlein Purpura

Henoch-Schönlein Purpura

- **Pathophysiology**: secondary to IgA immune complex deposition
- **Etiology**: often occurs following an upper respiratory tract infection, which leads to generation of IgA
- The **classic clinical triad** of Henoch-Schönlein purpura is
  - Palpable purpura on the buttocks and legs (100% of cases)
  - Arthralgias (75-85% of cases); usually localized to the knees and ankles, more in the lower limbs and oligoarticular
  - Abdominal pain
- **The most common renal manifestation**: Hematuria (20-55% of cases)
- **The most common GI complication**: Ileoileal intussusception
- **Management**: Disease is self-limited; treat with steroids if severe

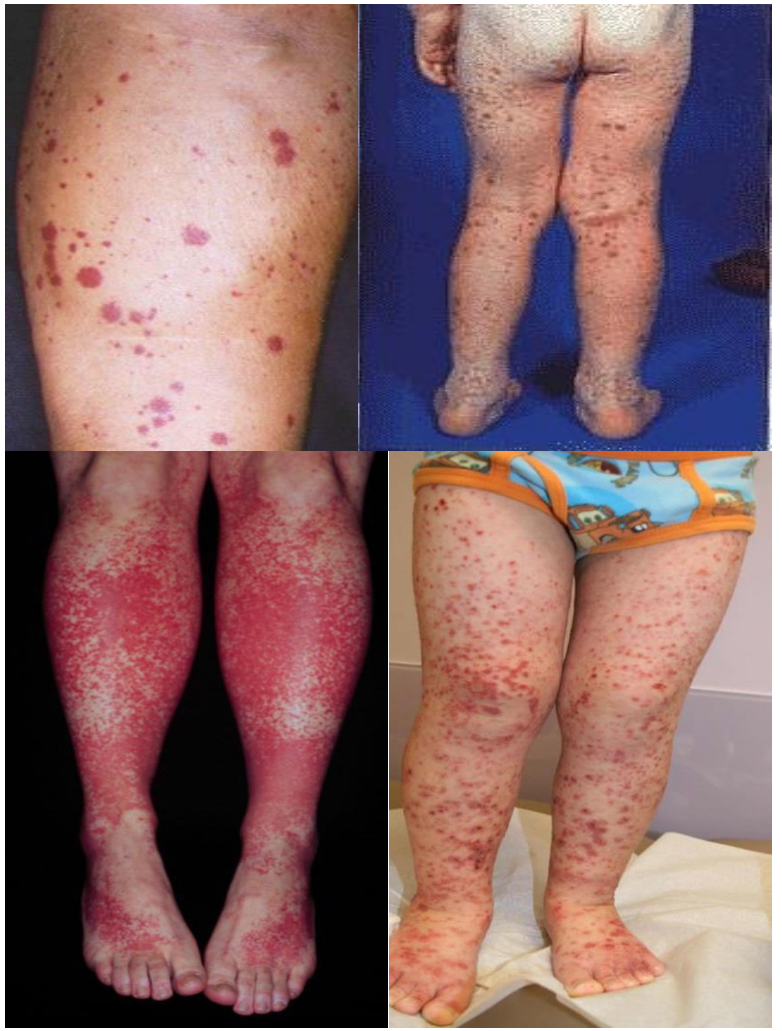
4-year-old patient presented with rash, abdominal pain and ankle pain



- **What is the diagnosis ?**
  - Henoch-Schönlein Purpura
- **Mention 2 other clinical findings in this patient**
  - Arthralgia
  - Abdominal pain
- **Other DDx**
  - ITP, Protein S or C deficiency
- **Mention 2 needed investigation to diagnose**
  - HSP is clinically diagnosed
  - Urinalysis showing hematuria
  - Renal and/or skin biopsy

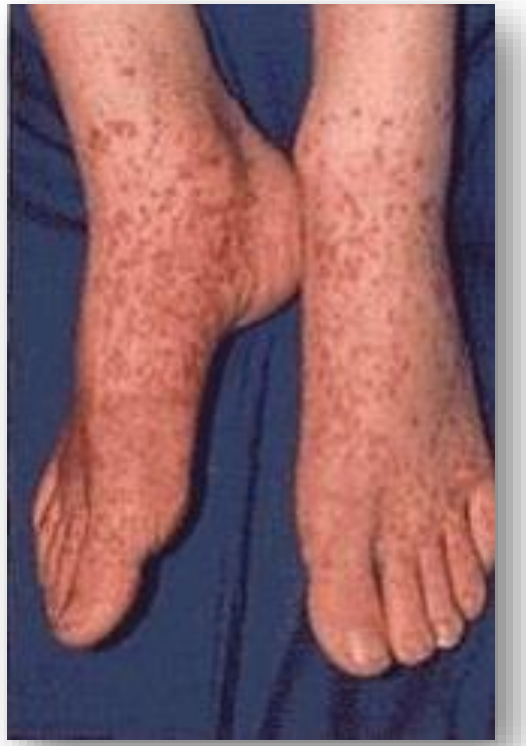
Henoch-Schönlein Purpura

- **What's your Dx.?**
  - Henoch-Schönlein Purpura
- **Mention other 3 symptoms**
  - Abdominal pain
  - Arthralgia
  - Renal manifestation (hematuria)
- **If the patient has a toxic appearance with a rapidly evolving rash. What will be your most probable Dx. ?**
  - Acute Meningococcemia.
- **What other body organs you will examine ?**
  - Joints, abdomen, eyes.
- **Give 2 important lab tests you will order.**
  - KFT, Skin biopsy from the lesion, Urine analysis.



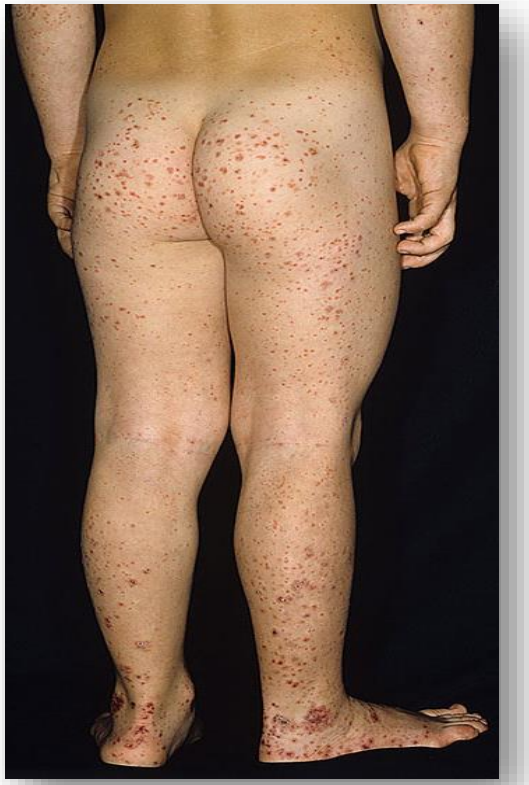
Child complains of abdominal pain and limping

- **What is the diagnosis ?**
  - Henoch-Schönlein Purpura
- **Skin manifestation percentage**
  - 100%
- **What is the histological finding ?**
  - IgA deposition
- **True or false, progression to end stage renal disease is 30%**
  - False



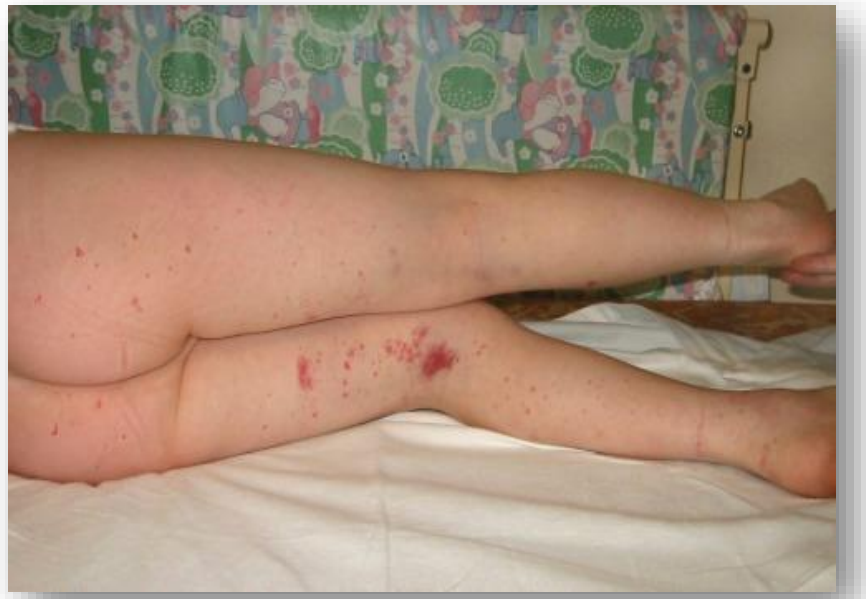
Henoch-Schönlein Purpura

- **What's the name of rash ?**
  - Purpuric rash
- **Give two other symptom ?**
  - Abdominal pain, arthralgia
- **Name four possible complications of this condition**
  - **GI**: Intussusception + Hepatosplenomegaly + bowel perforation
  - **GU**: renal involvement with development of the nephrotic syndrome. + An infrequent complication of scrotal edema is testicular torsion, which may be suggested by pain and must be treated promptly
  - **HLS**: Lymphadenopathy
  - **CNS**: "A rare but potentially serious outcome of central nervous system (CNS) involvement is the development of seizures, paresis, or coma."



Henoch-Schönlein Purpura

- A 7-year-old boy presented with intermittent abdominal pain and difficulty walking due to painful knees. He had been previously well except for a mild upper respiratory tract infection approximately one week ago. On examination he seems well but a non-blanching rash was seen on the buttocks and the legs (see photo). The rest of the body was spared



- **What is the diagnosis ?**
  - Henoch-Schönlein Purpura (HSP)
- **What renal complication can occur ?**
  - Glomerulonephritis

إضافي

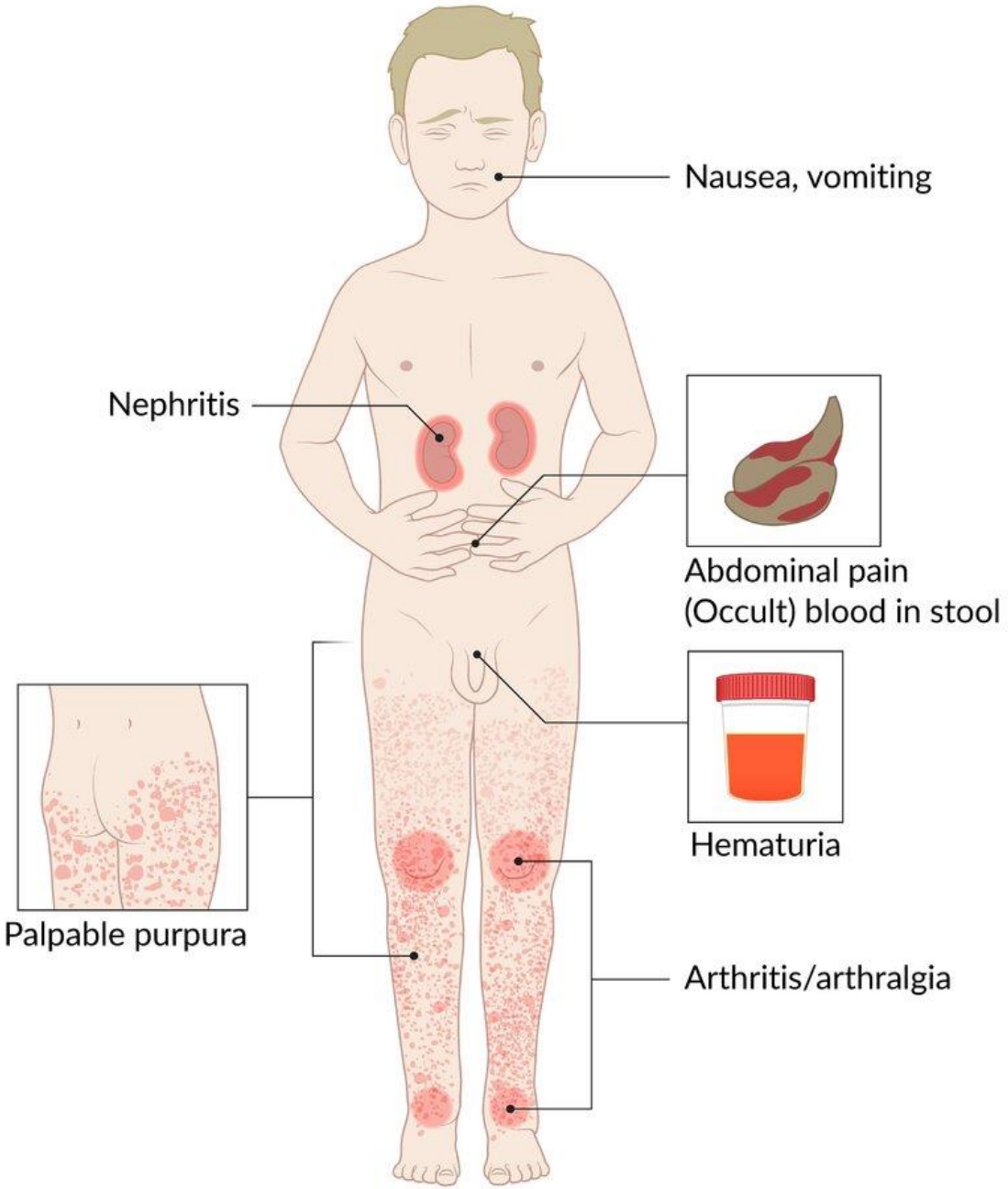
IgA vasculitis

Etiology
Often triggered by upper respiratory tract infection (causes IgA immune complex deposition in small vessel walls)

Diagnostics
Primarily clinical diagnosis

Treatment
Most cases are self-limiting
Mild disease: supportive treatment (e.g., NSAIDs)
Severe disease: systemic glucocorticoids

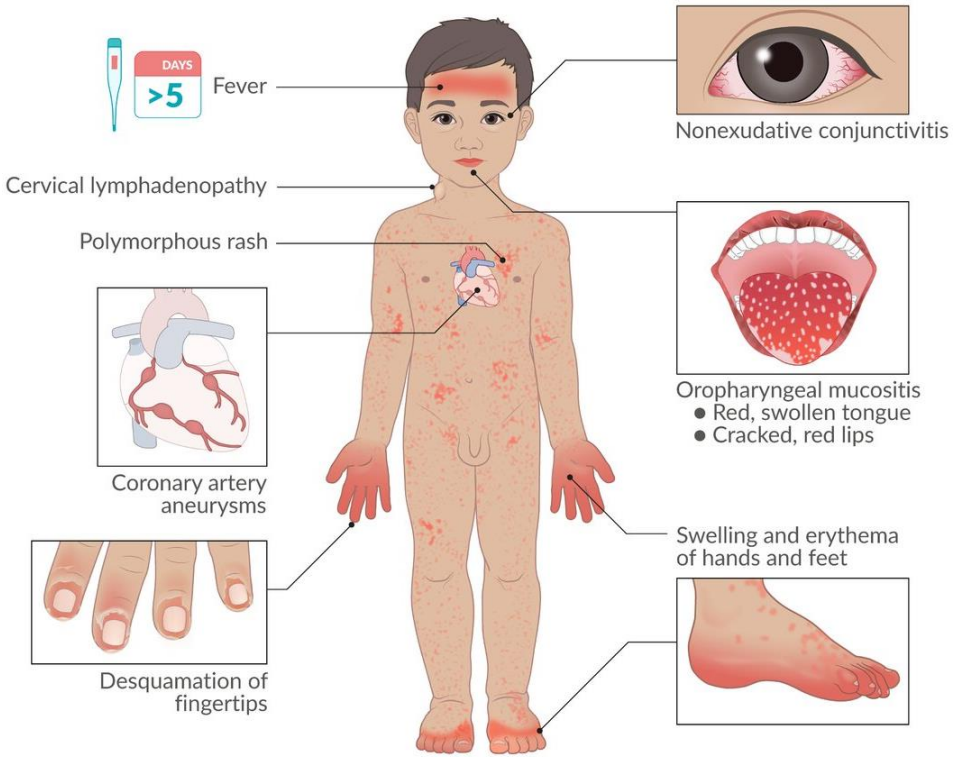
Complications
GI: intussusception, bowel ischemia/perforation
Renal: progressive kidney involvement (e.g., nephrotic syndrome), chronic kidney disease



Kawasaki

Kawasaki disease

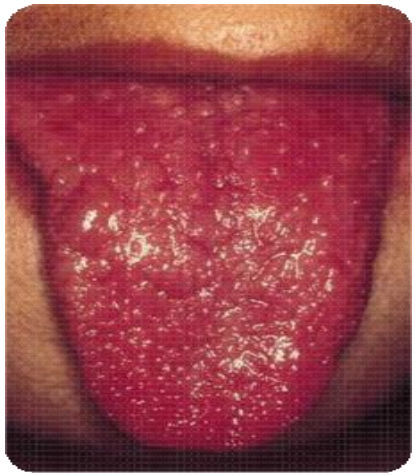
- The symptoms of Kawasaki disease may be remembered with the mnemonic "**CRASH & burn**":
  - **C**: **C**onjunctivitis
  - **R**: **R**ash (polymorphous -> desquamating)
  - **A**: **A**denopathy (cervical lymph nodes)
  - **S**: **S**trawberry tongue (oral mucositis)
  - **H**: **H**and-foot erythema and edema
  - **&**
  - **Burn**: Fever

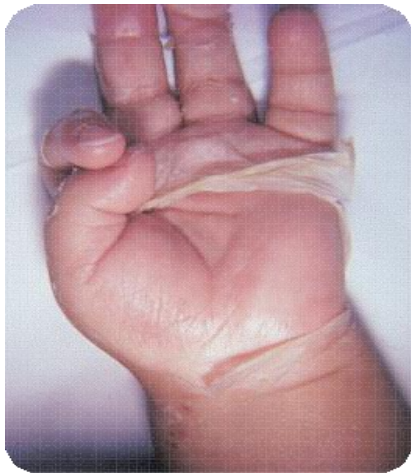


Kawasaki disease

❖**What is the criteria for the diagnosis of this disease?**

- Fever > 5 days & 4 out of 5:
  1. Non-purulent bulbar Conjunctivitis
  2. Polymorphous Rash
  3. Cervical lymphAdenitis
  4. Changes in the lips and mucus membranes (Strawberry tongue)
  5. Extremity (Hand & foot) skin changes (redness, swelling, peeling of the skin)





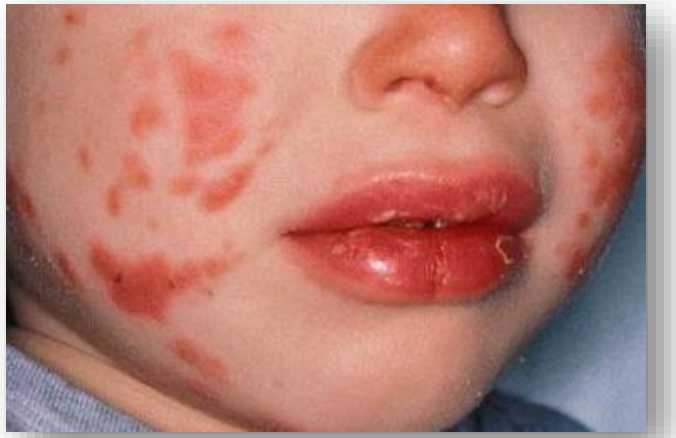
What's the name of each sign in the pictures ?

A. Non-exudative bilateral conjunctivitis

B. Desquamation

C. Strawberry tongue

A child present with fever for 1 week, and conjunctivitis



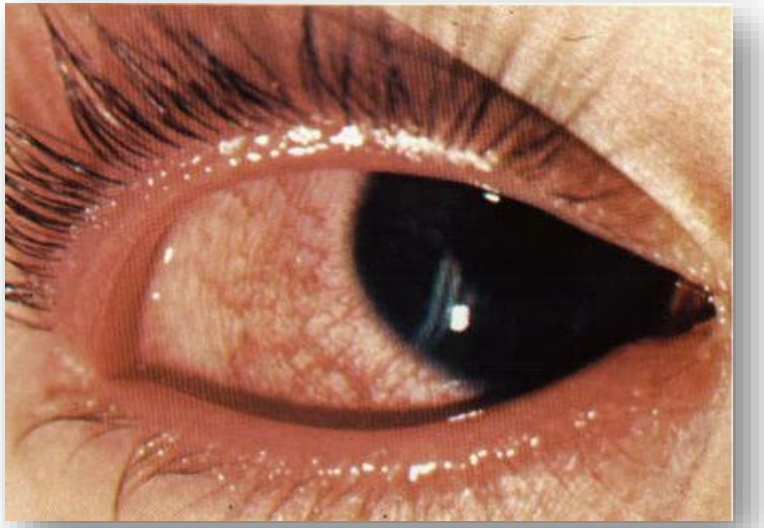
- **What is the diagnosis ?**
  - Kawasaki disease
- **Write down 2 modalities of treatment of this patients?** [سنوات (3)]
  - IVIG , aspirin
- **What are feared complications in this disease ?**
  - Coronary artery aneurysms, which can rupture; therefore, get an echo if Kawasaki is suspected
  - Coronary artery thrombosis, which can lead to myocardial infarction
  - Myocarditis

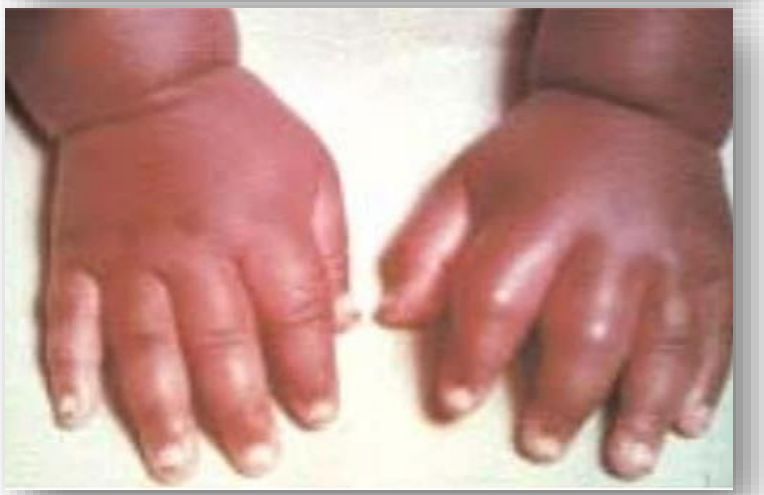
Kawasaki disease

- 4-year-old patient has fever for 1 week duration with polymorphous rash.

- **What is the signs seen in the pictures ?**
  - Conjunctival non-suppurative injection
  - Indurated (edema) and erythema of the hands

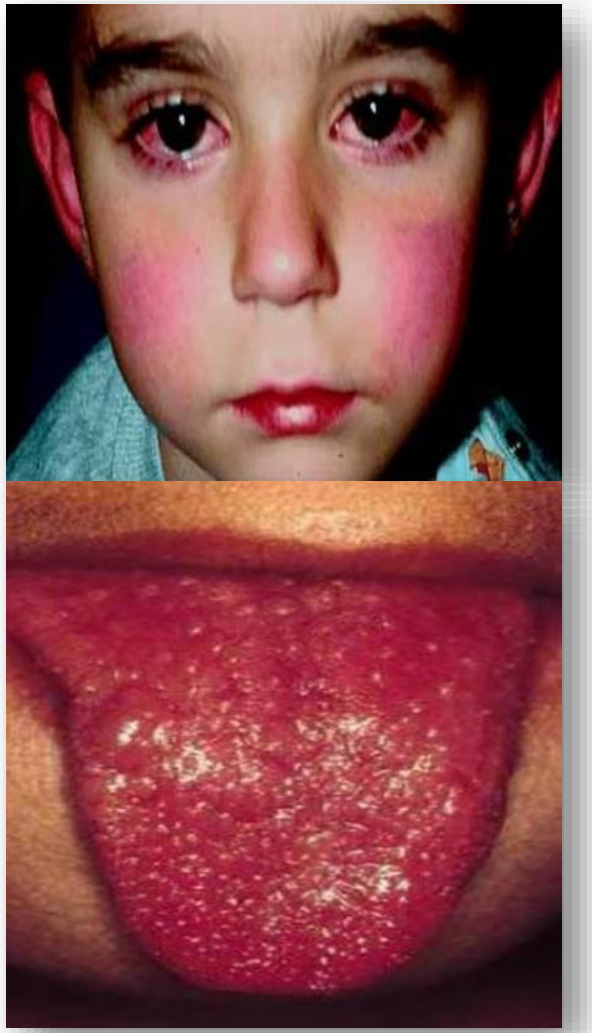
- **What is the serious complication could occur in this patient ?**
  - Coronary artery aneurysm





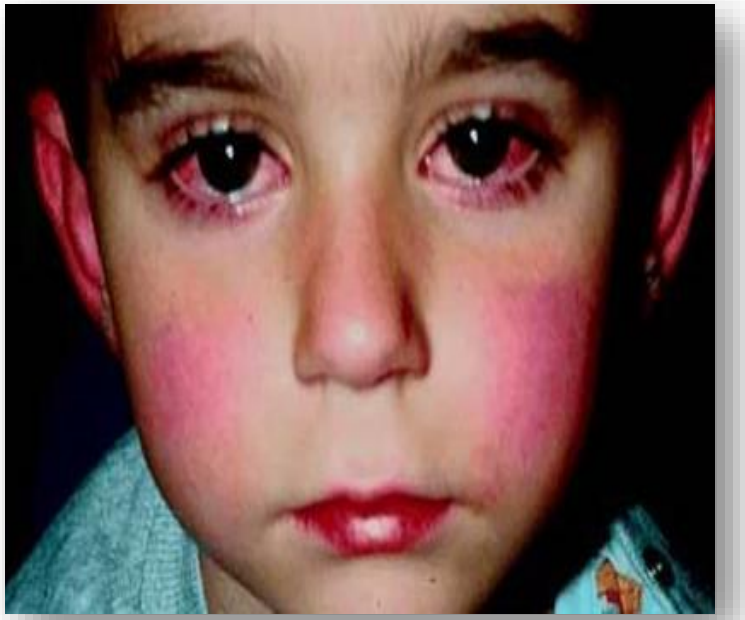
Child present with 6 days fever

- **What's Your Dx.?**
  - Kawasaki Disease.
- **Mention 2 other organs you want to examine.**
  - Hands & feet (peeling), Trunk (rash), Cervical lymph nodes.
- **Name the sign observed at the eyes.**
  - Non suppurative conjunctivitis.
- **What is the most serious complication?**
  - Inflammation of coronary arteries.



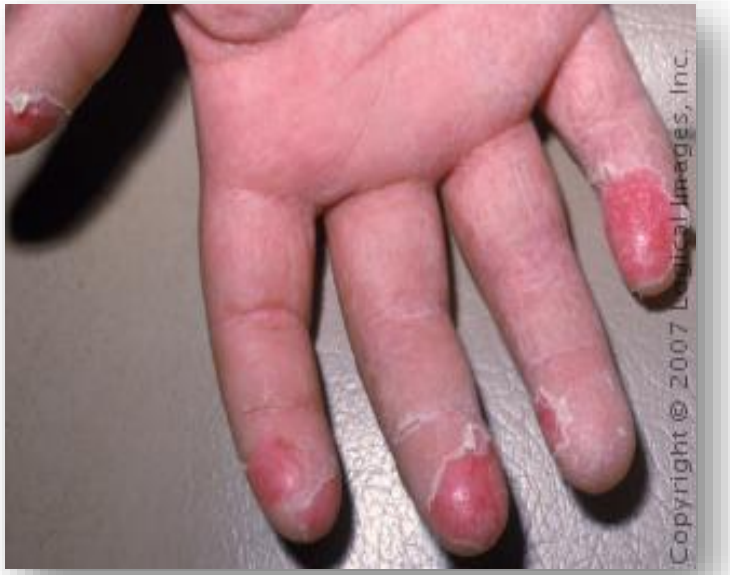
Kawasaki disease

- **Name two differential diagnosis for the child's eye condition?**
  - Kawasaki disease
  - Measles
  - Scarlet fever
- **What lab findings rule out Kawasaki ?**
  - KD is unlikely if the ESR, CRP, and platelet counts are normal after 7 days of fever



Kawasaki disease

- **Which this stage ?**
  - Subacute phase
- **What other thing we can see in this stage ?**
  - Thrombocytosis
  - Development of coronary artery aneurysm
  - Sudden death in patients who develop aneurysms
- **How to monitor the development of coronary artery aneurysm in Kawasaki ?**
  - Two-dimensional echocardiography
- **Mention a side effect for treatment ?**
  - Aspirin: Increase risk of bleeding, Hypertension



Clinical phases of Kawasaki disease next slide

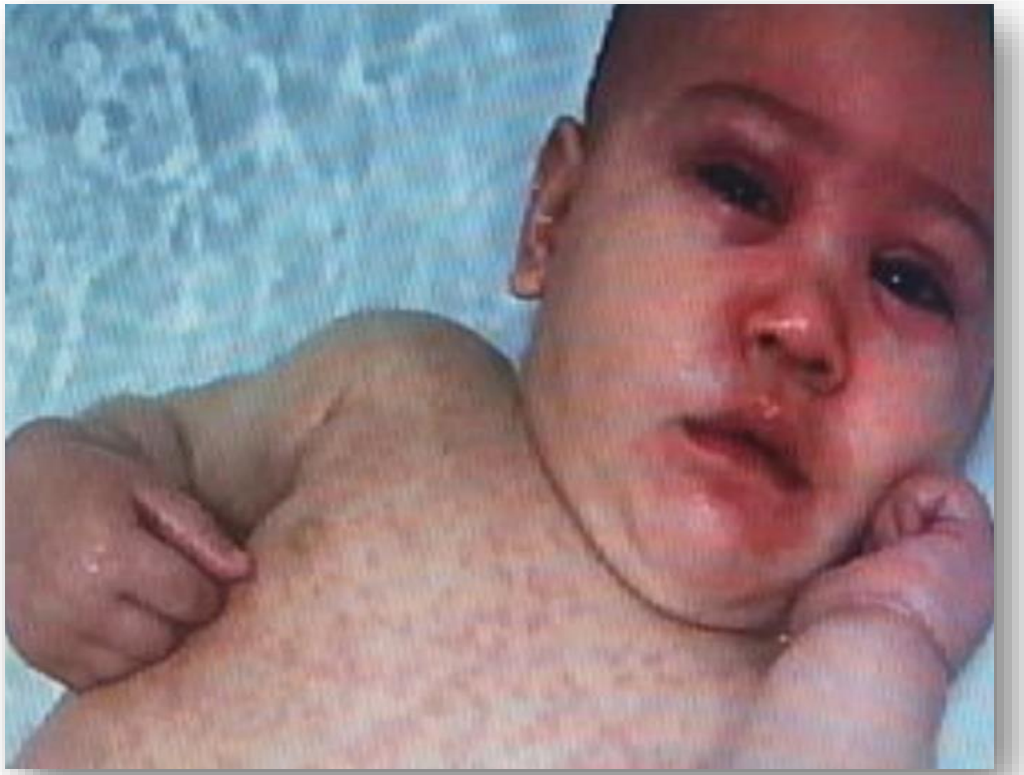
Clinical phases of Kawasaki disease

Acute phase	Subacute phase	Convalescent phase
CRASH & Burn • **C**: **C**onjunctivitis • **R**: **R**ash • **A**: **A**denopathy • **S**: **S**trawberry tongue • **H**: **H**and-foot erythema and edema • **&** • **Burn**: Fever	**Associated with** • Desquamation • Thrombocytosis • Development of coronary artery aneurysm • The highest risk of sudden death in patients who develop aneurysms	Begins when all clinical signs of illness have disappeared and continues until the erythrocyte sedimentation rate (ESR) returns to normal
Usually lasts 1-2 weeks	Generally, lasts 3 weeks	Typically, 6-8 weeks

9-month-old male, temperature 40, ESR 40 & the following rash

❖**The most likely diagnosis is**

a. Kawasaki
b. Scarlet fever
c. Measles
d. Roseola infantum
e. Hand foot mouth disease



Kawasaki disease

- A child presented with fever for 1 week
- **Give 2 lab test to help in diagnosis:**
  - ESR, CRP, CBC
- **What other sites you would like to examine:**
  - Mouth, hands and feet, heart

إضافي

"Defining Kawasaki disease and pediatric inflammatory multisystem syndrome-temporally associated to SARS-CoV-2 infection" key takeaways:

1. Children with Kawasaki disease-like multisystemic disease were significantly older at onset and presented more frequently from gastrointestinal and respiratory involvement.
2. Cardiac involvement was more common in patients with Kawasaki disease-like multisystemic disease, with 60.4% demonstrating myocarditis.
3. The risk of ICU admission was higher patients with Kawasaki disease-like multisystemic disease.
4. Coronary artery abnormalities were more common in children with Kawasaki disease.
5. COVID-19 positivity was prevalent in 75.5% of children in the Kawasaki disease-like multisystemic disease group, compared with 20% among those with Kawasaki disease ($P < .0001$).

Infectious exanthem

Infectious exanthem

Name	Number	Virus	Rash
Measles (Rubeola)	First disease	Measles virus	Erythematous macules and papules appearing first on the head and spread down over body over 3 days.
Scarlet fever	Second disease		The rash is red and feels like sandpaper and the tongue may be red and bumpy
Rubella (German measles)	Third disease	Rubella virus	Pink macules and papules that appear first on the head and spread down over body in 24 hours.
Erythema infectiosum	Fifth disease	Parvovirus B19	Confluent erythematous and edematous patches on cheeks ("slapped cheek") for 1-4 days followed by a "lacy," reticular, erythematous rash on the body.
Roseola infantum	Sixth disease	HHV-6 & HHV-7	Rapid onset of erythematous, blanching macules and papules surrounded by white halos on the trunk after 3-5 days of high fever. The rash spreads to the neck and body extremities and lasts 1-2 days.
Hand-foot-and-mouth disease	-	Coxsackie A virus	Macular rash limited to hands, feet and the mouth

Fourth disease, also known as "Dukes' disease" is a condition whose existence is not widely accepted today.

Measles

- **Family**: Paramyxoviridae (negative sense RNA viruses, enveloped)
- **Route of transmission**: Respiratory droplets
- The **4 Cs** of Measles
  - **C**ough
  - **C**oryza (runny/stuffy nose due to inflammation)
  - **C**onjunctivitis
  - **K**oplik spots (bright red spots with a(n) blue-white center on buccal mucosa)
- **Exanthem**: Maculopapular rash that develops 1-2 days following the prodromal symptoms of measles; starts on the face and moves down-wards
- **Management**: Supportive: fluids /antipyretics, Vitamin A
- **Prevention**: MMR (Live, attenuated) vaccine at 9, 12, 18 months
- **Serious complication**: Subacute sclerosing panencephalitis

Measles

- 18-month-old child with rash, runny nose and conjunctivitis

❖ **Mention 2 ddx**

- Measles and Kawasaki

❖ **Distribution of rash**

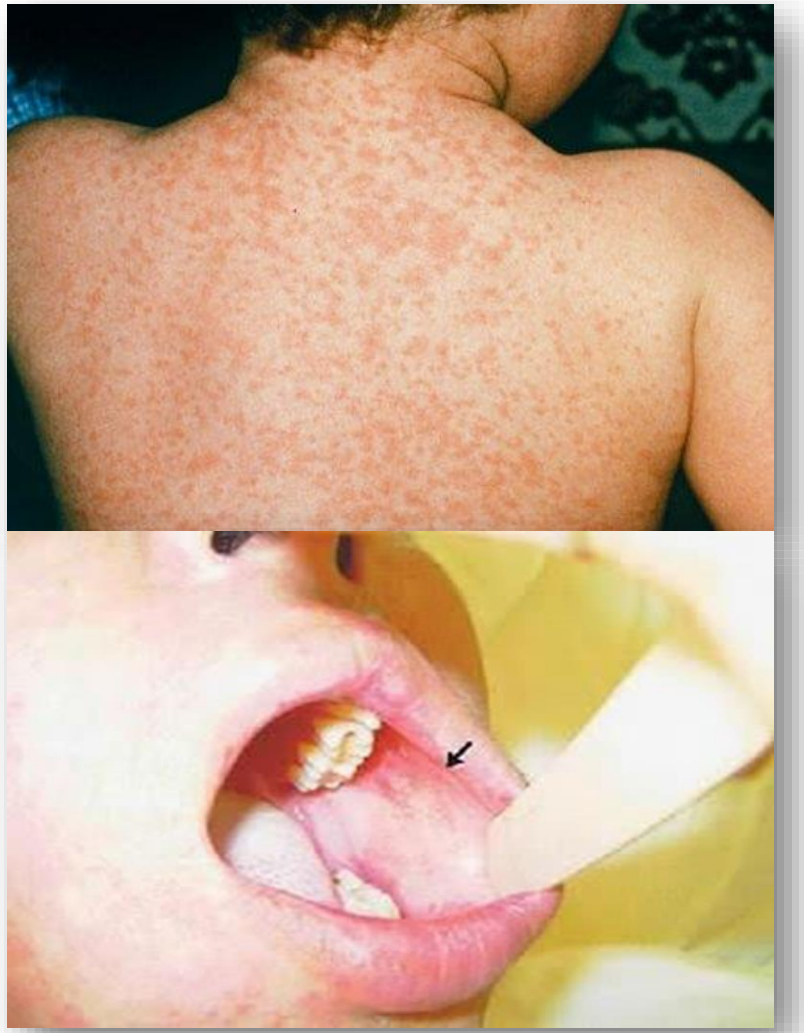
- cephalocaudal and centrifugally

❖ **Vaccines in Jordan at what age ?**

- 9, 12, 18 months

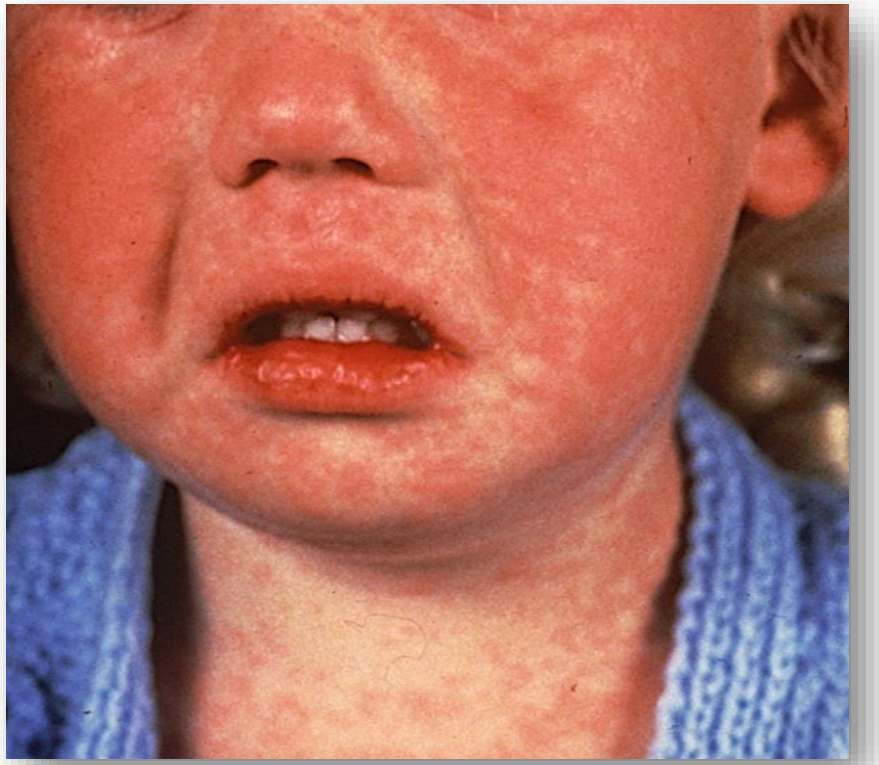
❖ **What is seen in the second picture ?**

- Koplik spots (bright red spots with a blue-white center on buccal mucosa)



Measles

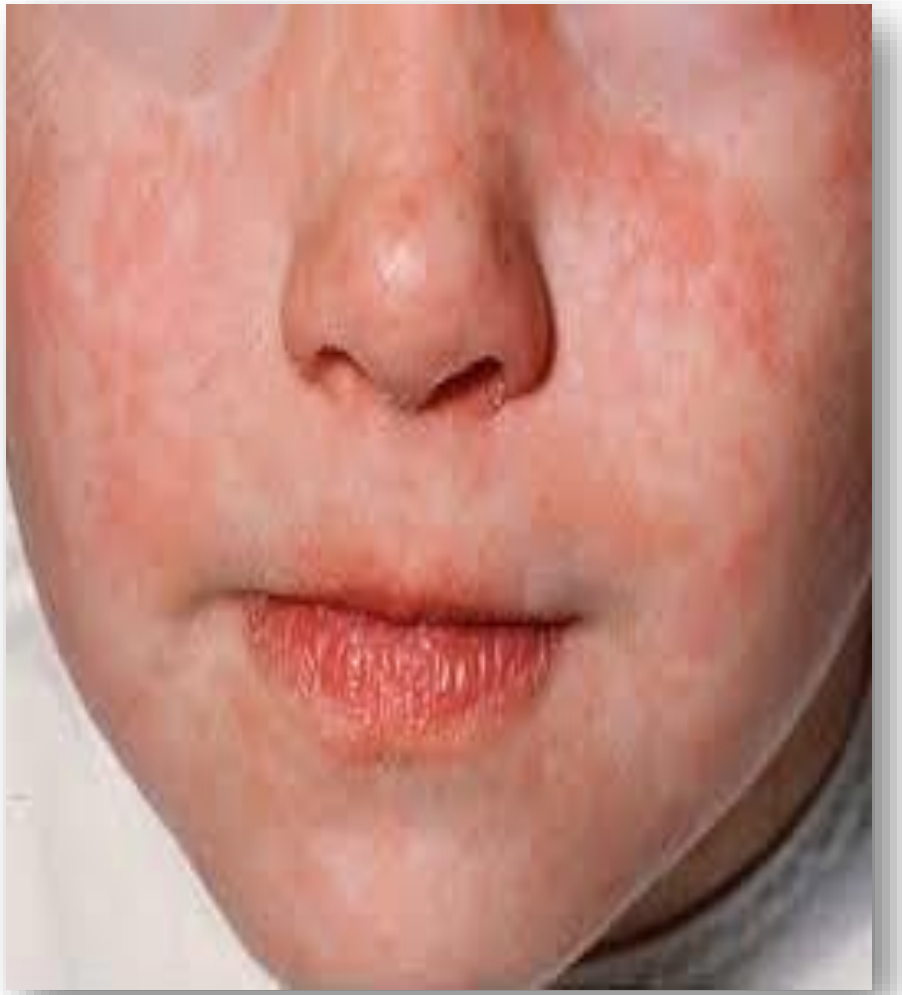
- **What is your diagnosis ?**
  - Measles
- **When is the vaccine given for this rash?**
  - 9, 12, 18 months



Measles

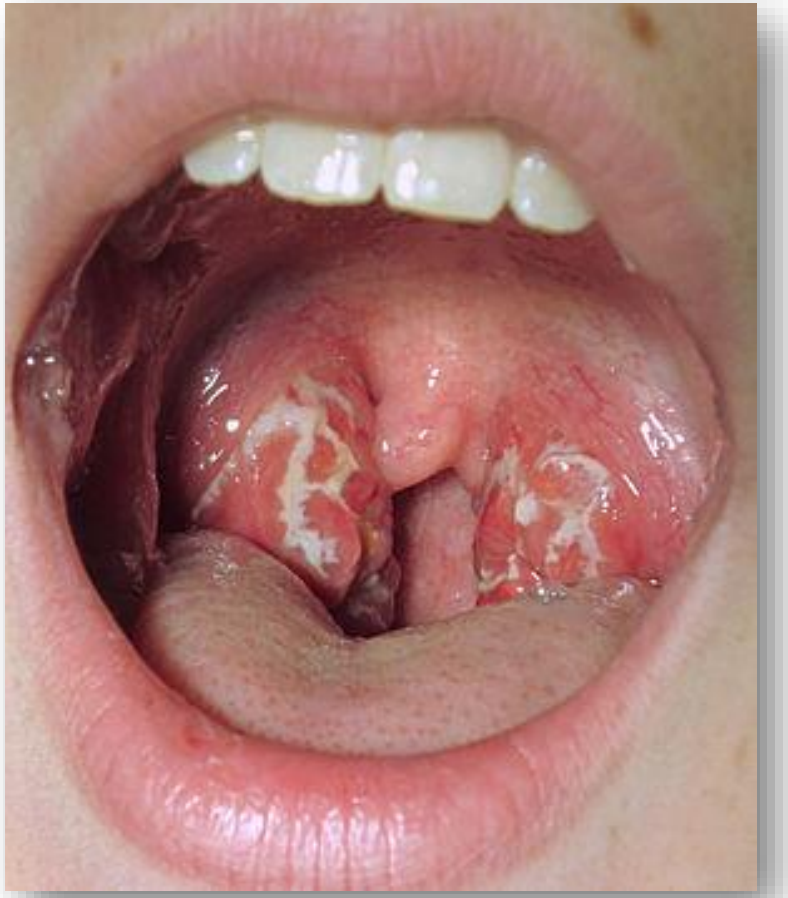
- 1 year old child comes with fever 3 days duration, coryza, skin rash and conjunctivitis

- **Give 3 questions you would ask in hx**
  - Vaccine hx
  - distribution of rash
  - The duration of fever and rash
- **Management**
  - Supportive: fluids /antipyretics
  - Vitamin A



Scarlet fever

- **What is the most common organism ?**
  - Group A streptococci
- **Toxin mediated disease of that organism ?**
  - Scarlet fever (Erythrogenic exotoxin (SpeA, B, C))
  - Toxic shock like syndrome (SpeA and C)
  - Necrotizing fasciitis (SpeB)
- **Mention an immunomediated disease of that organism ?**
  - Rheumatic fever
  - Post-streptococcal glomerulonephritis



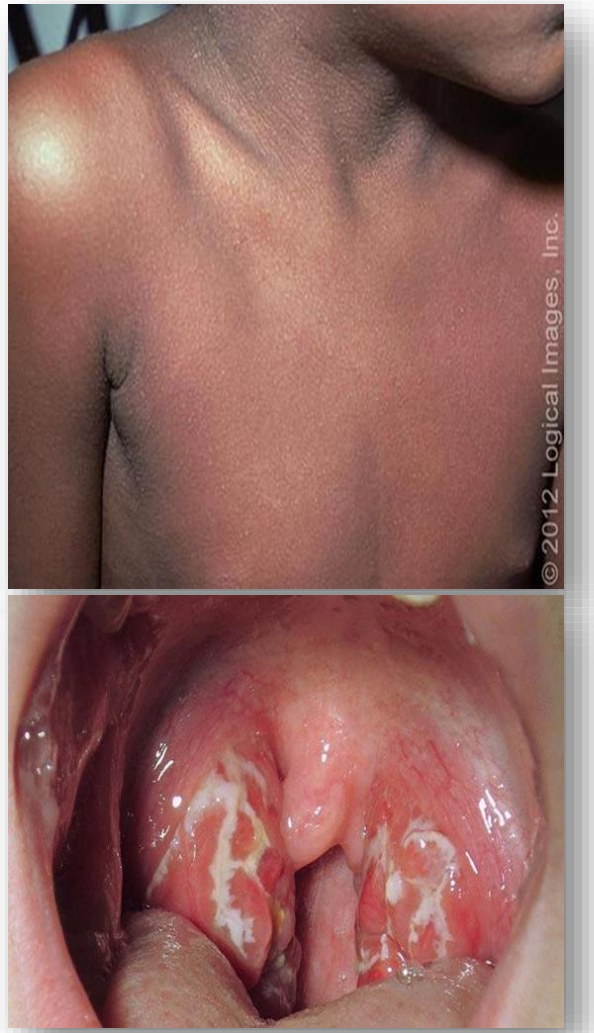
➢ Scarlet fever can present with a red, swollen, "strawberry" tongue, pharyngitis, and a diffuse, "sandpaper-like" rash that spares the face

Scarlet fever

- **What is the diagnosis ?**
  - Scarlet fever
- **Mention 2 immunological complications of that organism ?**
  - Rheumatic fever
  - Post-streptococcal glomerulonephritis

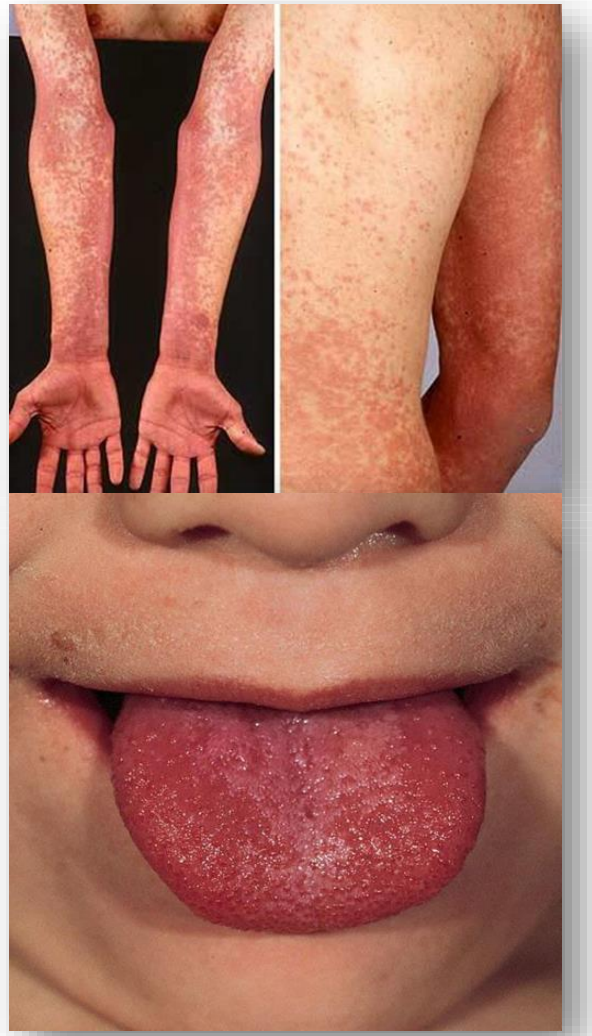
A patient presents with sandpaper like rash & sore throat.

- **What is the most common organism ?**
  - Group A streptococci
- **Mention 2 complications of that organism ?**
  - Rheumatic fever
  - Post-streptococcal glomerulonephritis



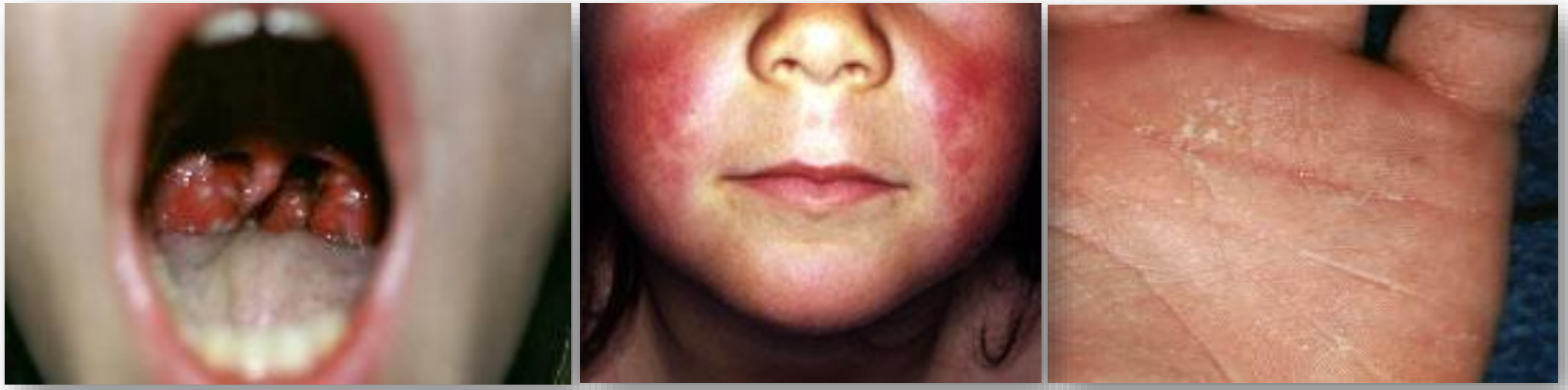
Scarlet fever

- This patient presented with history of sore throat and fever 3 weeks ago and this rash
- **What is the causative microorganism?**
  - Group A strep
- **What's the Diagnosis ?**
  - Scarlet fever
- **What's the treatment ?**
  - Oral penicillin V (Note; the scarlet fever is caused by streptococcus pyogenes, GABHS).
  - In patients allergic to penicillin: macrolides



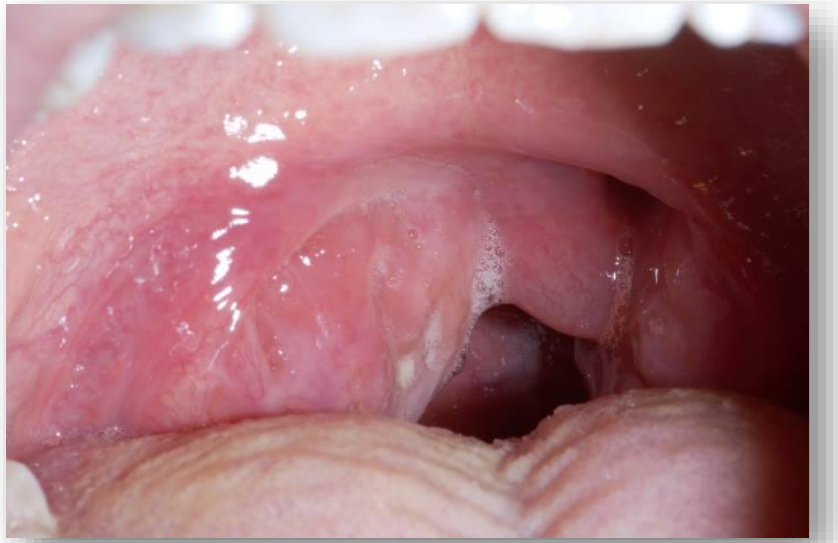
Scarlet fever

- **What's the Diagnosis ?**
  - Scarlet fever
- **Name 2 complications**
  - Peritonsillar abscess, sinusitis, bronchopneumonia and meningitis, or problems associated with immune system as rheumatic fever or glomerulonephritis.



Scarlet fever – Complications

- Patient present with trismus, previous history of pharyngotonsillitis
- **What are the findings ?**
  - Bulging of the right tonsil & deviation of uvula
- **What is your diagnosis ?**
  - Peri-tonsillar abscess
- **What is the treatment ?**
  - Drainage and IV Antibiotics

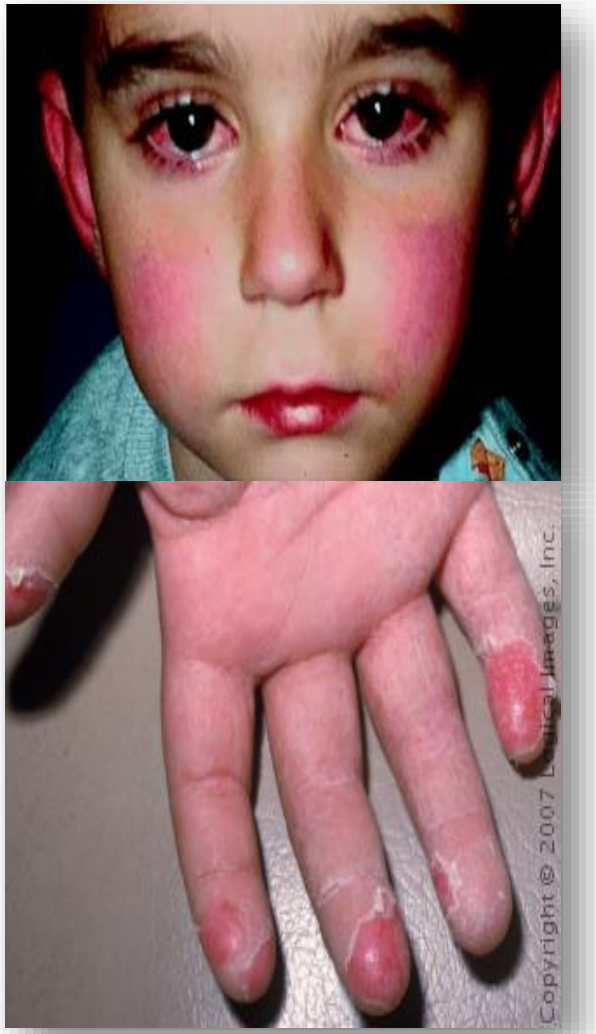


Scarlet fever

- **Mention 2 DDx ?**
  - Scarlet fever and Kawasaki disease
- **The organ which is affected by two DDx ?**
  - Heart

Mention 2 ddx with treatment for each one

- Kawasaki disease – IVIG, aspirin
- Scarlet fever – penicillin

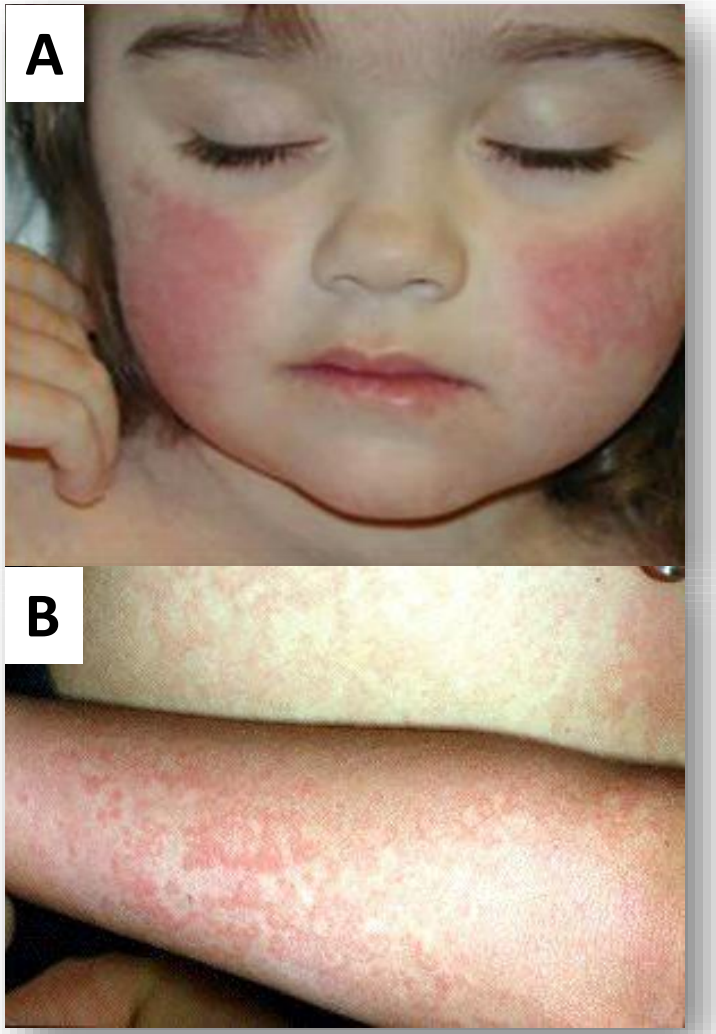


Erythema infectiosum

- **Causative agent**: parvovirus B19; Naked single-stranded DNA virus
- **Route of transmission**: Respiratory droplets, vertically
- **Exanthem**:
  - Rash that starts on the face and moves down-wards after a mild fever
  - Appearance: Slapped cheeks appearance, Lace-like rash
- **Management**:
  - Immunocompetent: Supportive
  - Immunocompromised: IVIG and RBC transfusion
- **Serious complication**:
  - Aplastic crisis in patients with sickle cell, thalassemia, and hereditary spherocytosis
  - Infection during the 1st and 2nd trimesters can result in hydrops fetalis
  - Infection in adults can result in small-joint arthritis, myocarditis, and edema

Erythema infectiosum

- **Describe what you see.**
  A. Slapped cheeks appearance.
  B. Lace-like rash
- **What is your diagnosis ?**
  - Erythema infectiosum
- **What is the causative organism ?**
  - Parvovirus B19
- **Write down three complications of this disease**
  - Aplastic anemia
  - Arthralgia / arthritis
  - Myocarditis

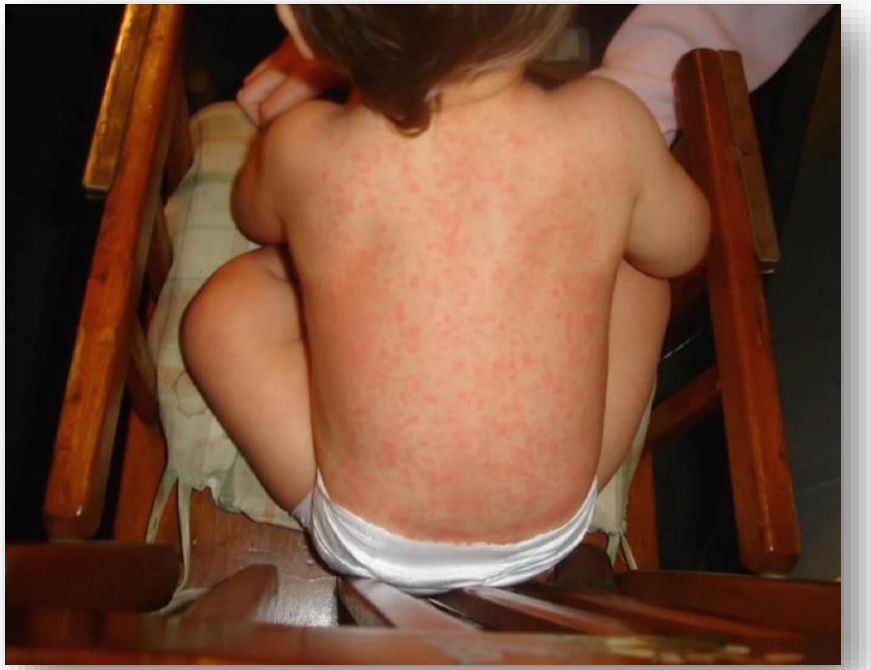


Roseola infantum (Exanthema subitum)

- **Causative agent**: HHV-6 and HHV-7; Herpesviridae family
- **Route of transmission**: unknown
- **Exanthem**:
  - Develops after a very high-grade fever that lasts 3-5 days
  - Starts on the neck/trunk and moves down-wards (doesn't affect the face)
  - Appearance: Red, "lacy" appearance
- **Management**: Supportive (infection is self-limiting)
- **Complication**: The most common cause of febrile seizures in children

Roseola infantum

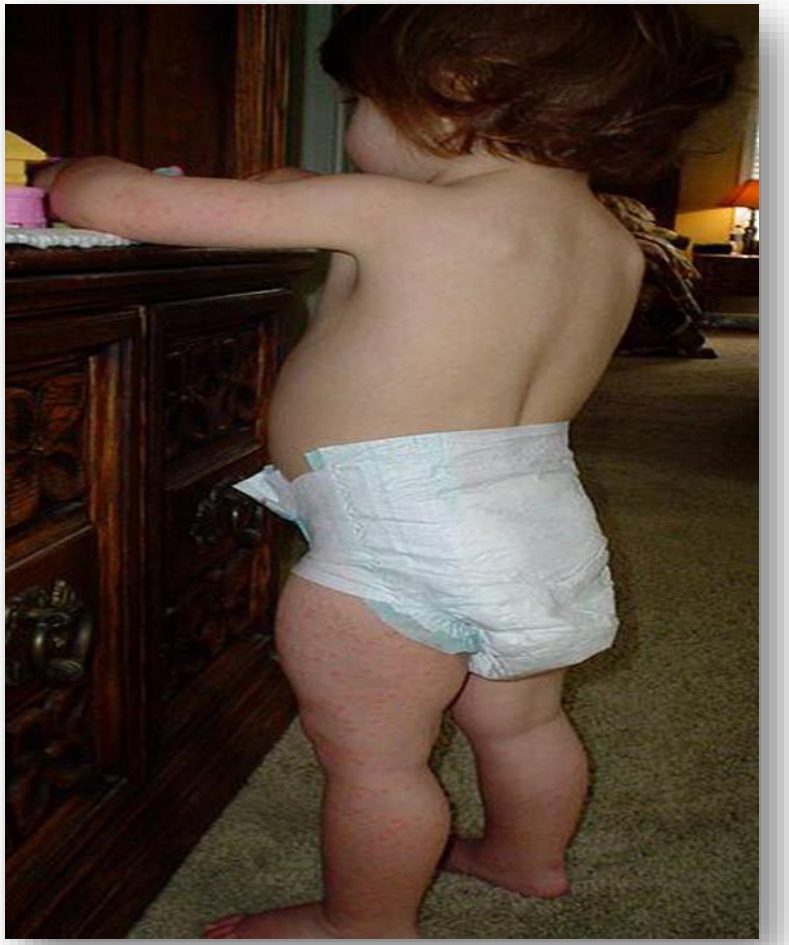
- Three days duration fever treated with amoxicillin and after that this rash appeared
- **What's your diagnosis ?**
  - Roseola infantum



Roseola infantum

- This child presented with a history of 3 days fever and upper respiratory symptoms the fever was documented as 40 degrees followed by this rash

- **What's your diagnosis ?**
  - Roseola infantum

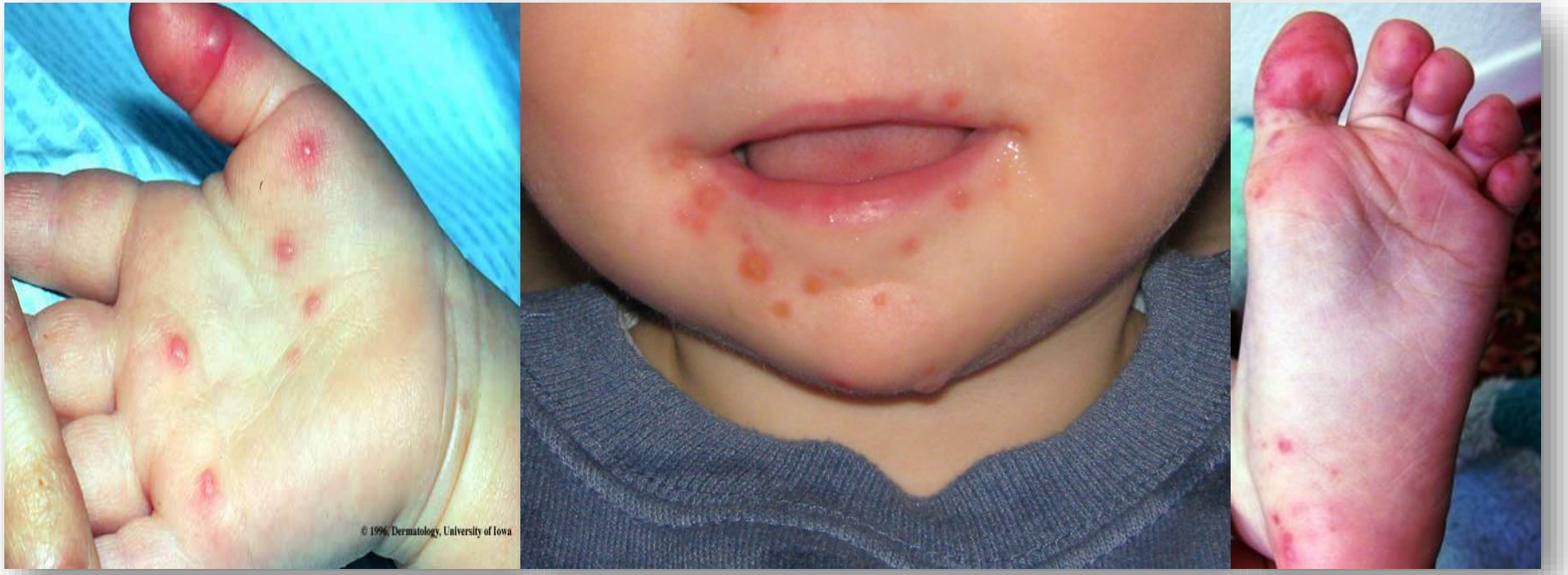


Hand-Foot-Mouth disease

- **Causative agent**: Coxsackie virus A; picornaviridae family
- **Route of transmission**: Fecal-oral
- **Exanthem**: Red, vesicular rash
- **Management**:
  - Adequate fluid intake
  - Antipyretics, Direct analgesia, Acetaminophen or ibuprofen
- **Complication**: Neurological sequelae; associated with the strain EV-71

Hand-Foot-Mouth disease

- **What is the diagnosis?**
  - Hand-Foot-Mouth disease
- **What is the causative micro-organism ?**
  - Coxsackie virus A

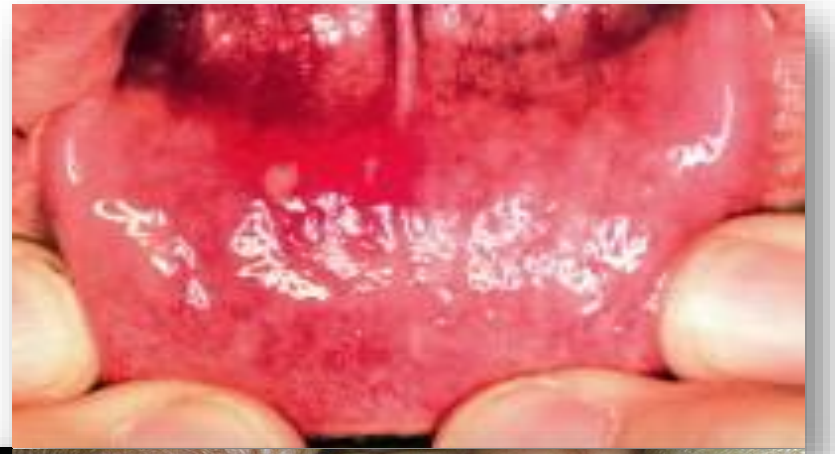


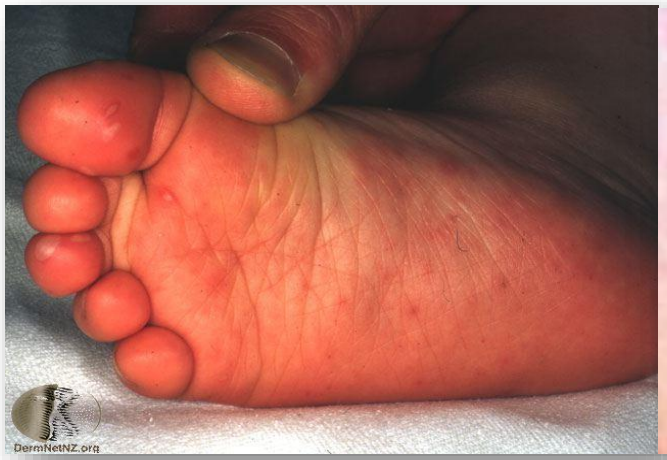
Hand-Foot-Mouth disease

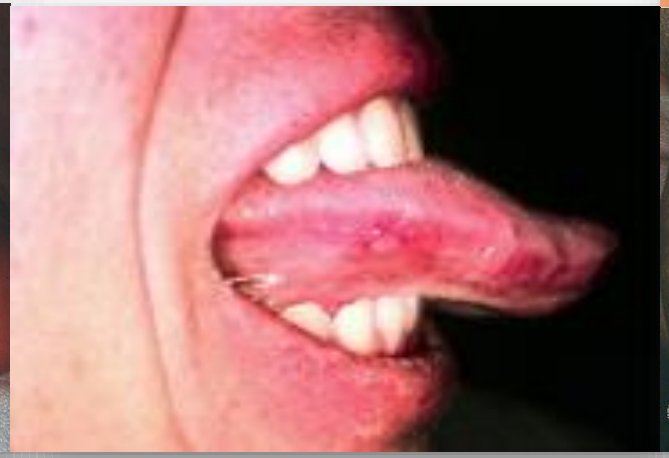
- **What is the diagnosis?**
  - Hand-Foot-Mouth disease
- **What is the causative micro-organism ?**
  - Coxsackie virus A

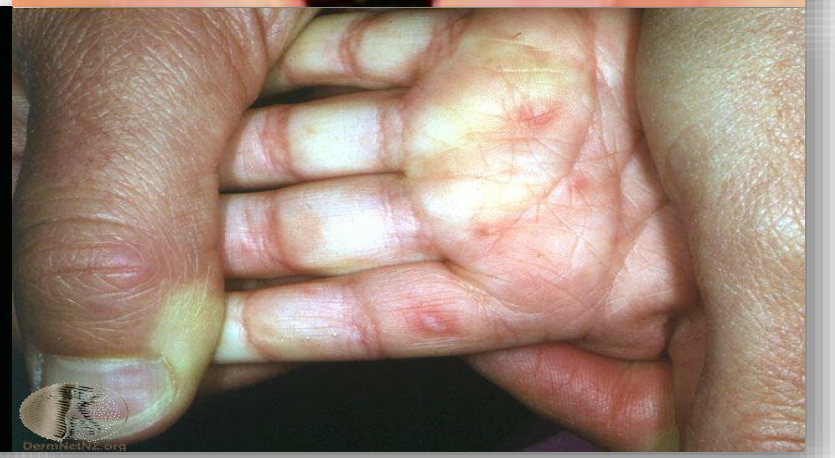
Hand-Foot-Mouth disease

- **What is the diagnosis?**
  - Hand-Foot-Mouth disease
- **What is the causative micro-organism ?**
  - Coxsackie virus A







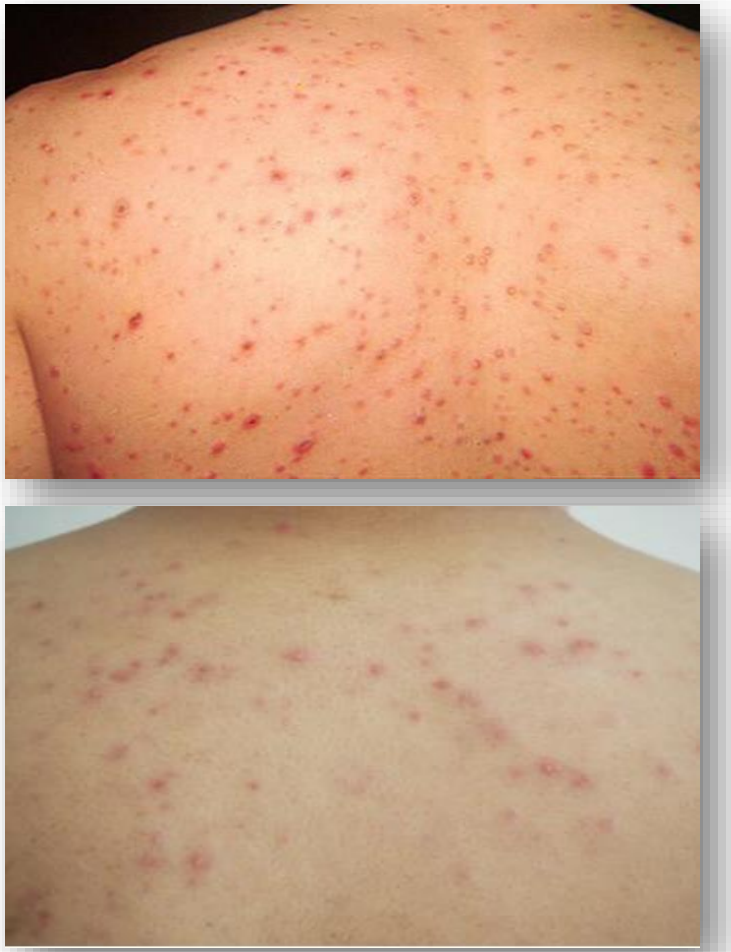


Chicken pox

- **Causative agent**: Varicella-Zoster Virus (HHV-3)
- **Route of transmission**: Respiratory droplets
- **Exanthem**: "dew drops on a rose" appearance
- **Management**: Supportive, Acyclovir if severe
- **Prevention**: Live, attenuated vaccine at 9 months
- **Complication**: Pneumonia, Encephalitis

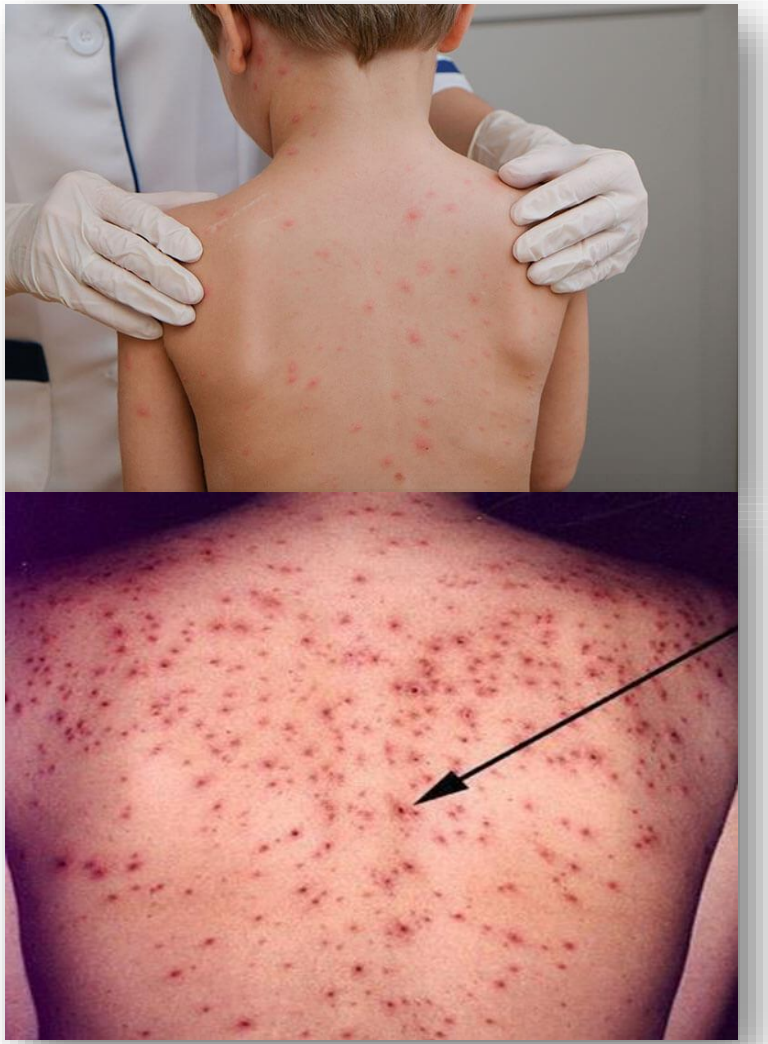
Chicken pox

- **What is the spot diagnosis?**
  - Chicken pox
- **What is the microbe causes of this condition?**
  - Varicella-zoster virus
- **Mention one complication**
  - Bacterial infection (cellulitis)
  - Pneumonia
  - Encephalitis
- **At which age should receive the vaccine**
  - First dose: 12- 15 months of age
  - Second dose: up to 4 years of age



Patient present with low grade fever

- **Type of vaccine**
  - Varicella > live attenuated
- **Time of vaccination**
  - First dose: 12- 15 months of age
  - Second dose: up to 4 years of age
- **Mention 2 complication**
  - Encephalitis , cellulites , pneumonia
- **Treatment if severe**
  - IV acyclovir



Chicken pox

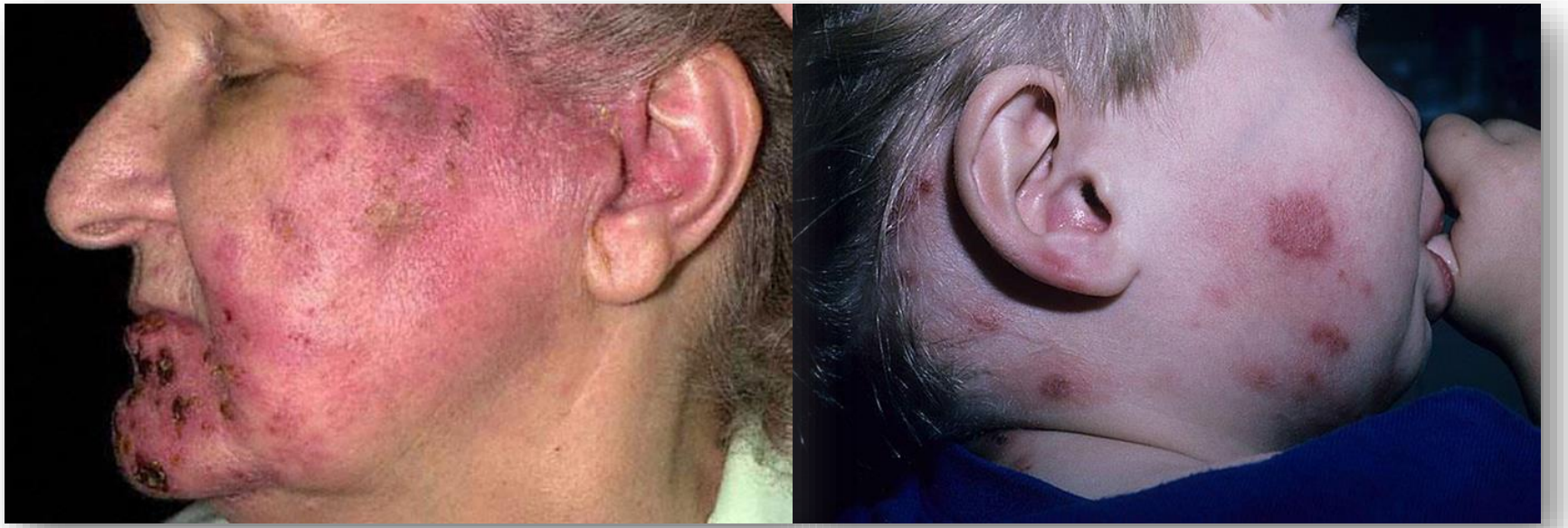
- **Your differential diagnosis ?**
  - Chicken pox
- **Describe the lesion ?**
  - Vesicles with different age and crust
- **What is your treatment ?**
  - Supportive

Chicken pox

- **What is the microbe causes of this condition?**
  - Varicella-zoster virus
- **What is the treatment if the patient is immunocompromised ?**
  - Intravenous acyclovir

Shingles

- **Name the disease.**
  - Shingles.
- **Mention 2 specific features for this disease.**
  - Dermatomal distribution, cluster of vesicles on an erythematous base.



Which of the following is not consistent clinical scenario for this picture

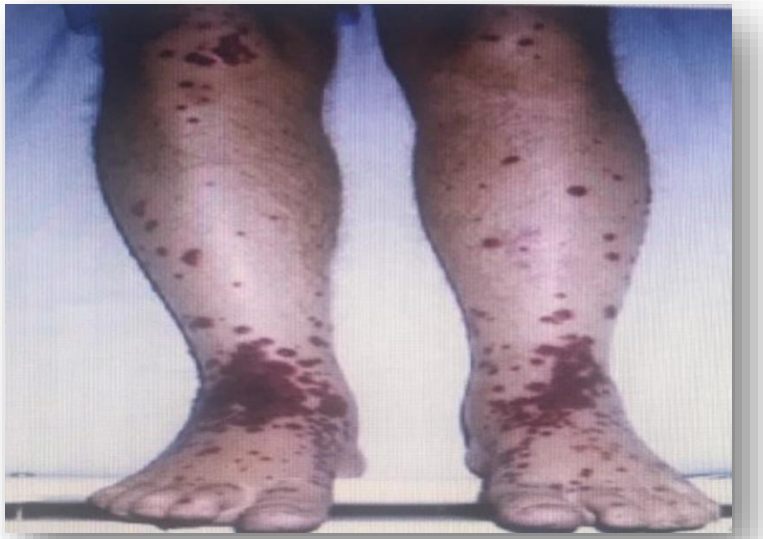
a. 15 years old female patient diagnosed 4 weeks ago with lupus

b. 6 years old male patient has hematuria and arthritis (Dx: HSP)

c. 2 years old unvaccinated complaining of septic shock and seizures (Dx: Meningococcemia)

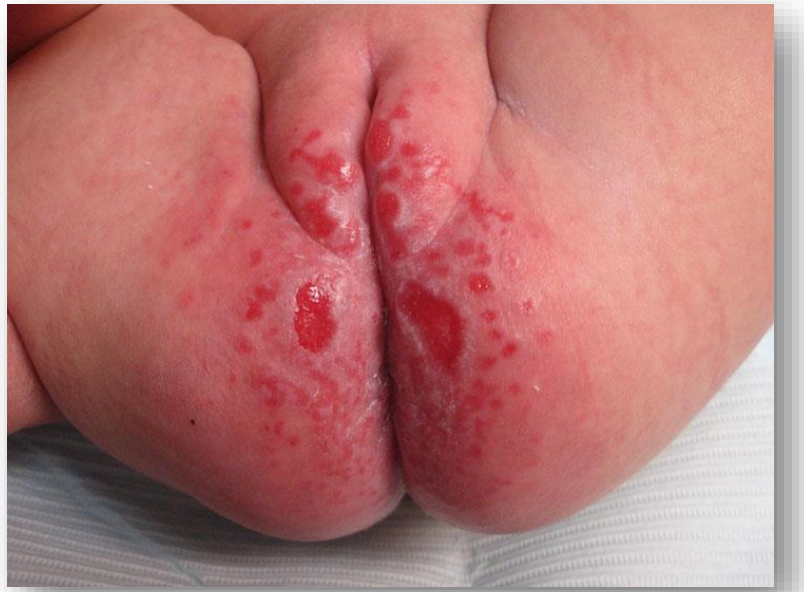
d. 4 years old male patient presents with severe sepsis admitted to the PICU

e. 2 years old male patient presented with kissing tonsils, hepatosplenomegaly and fever. (Dx: Mononucleosis)



Napkin rash

- **What's the name of this lesion ?**
  - Napkin rash
- **What's the treatment ?**
  - Something related to zinc
- **How to prevent ?**
  - Keep dry, frequent changing of diaper, using cream ……..



Oral thrush

- **What is this?**
  - Oral thrush secondary to Candida (mucosal candidiasis).
- **What's the cause ?**
  - Oral Candida/ fungal infection.
- **What is the treatment ?**
  - Topical nystatin

Acute rheumatic fever

Low-Yield

Acute rheumatic fever

- 6-year-old male patient presented with limping and the following skin lesion.

❖ **What is the name of this skin finding ?**

- Erythema marginatum

❖ **Name other 2 findings**

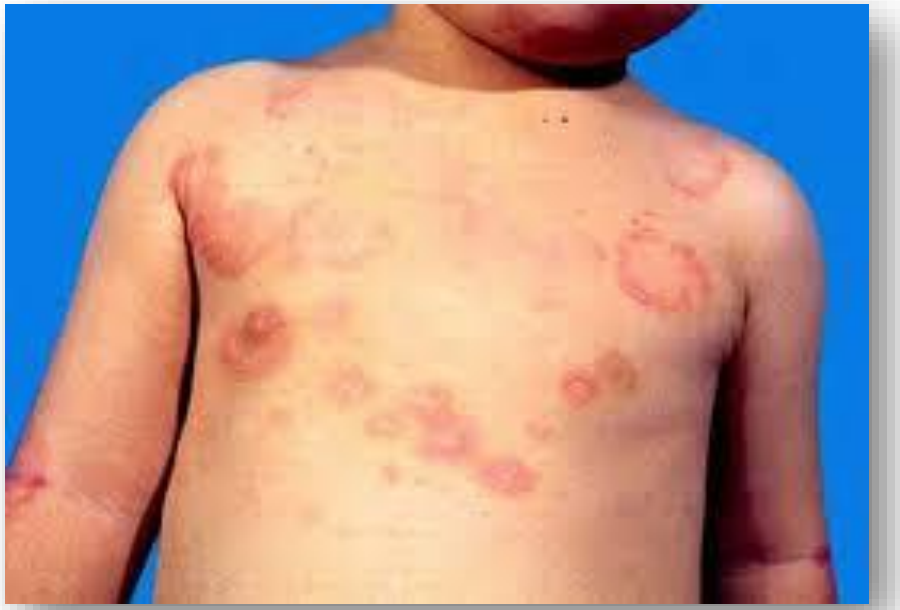
- Migratory polyarthritis, Carditis

❖ **What are the most likely diagnosis**

- Acute rheumatic fever

❖ **What are the investigation you need to confirm your diagnosis ?**

- ECG, Echocardiography



J♥NES (major criteria):
Joint (migratory polyarthritis)
♥ (carditis)
Nodules in skin (subcutaneous)
Erythema marginatum (evanescent rash with ring margin)
Sydenham chorea (involuntary irregular movements of limbs and face)

شرح

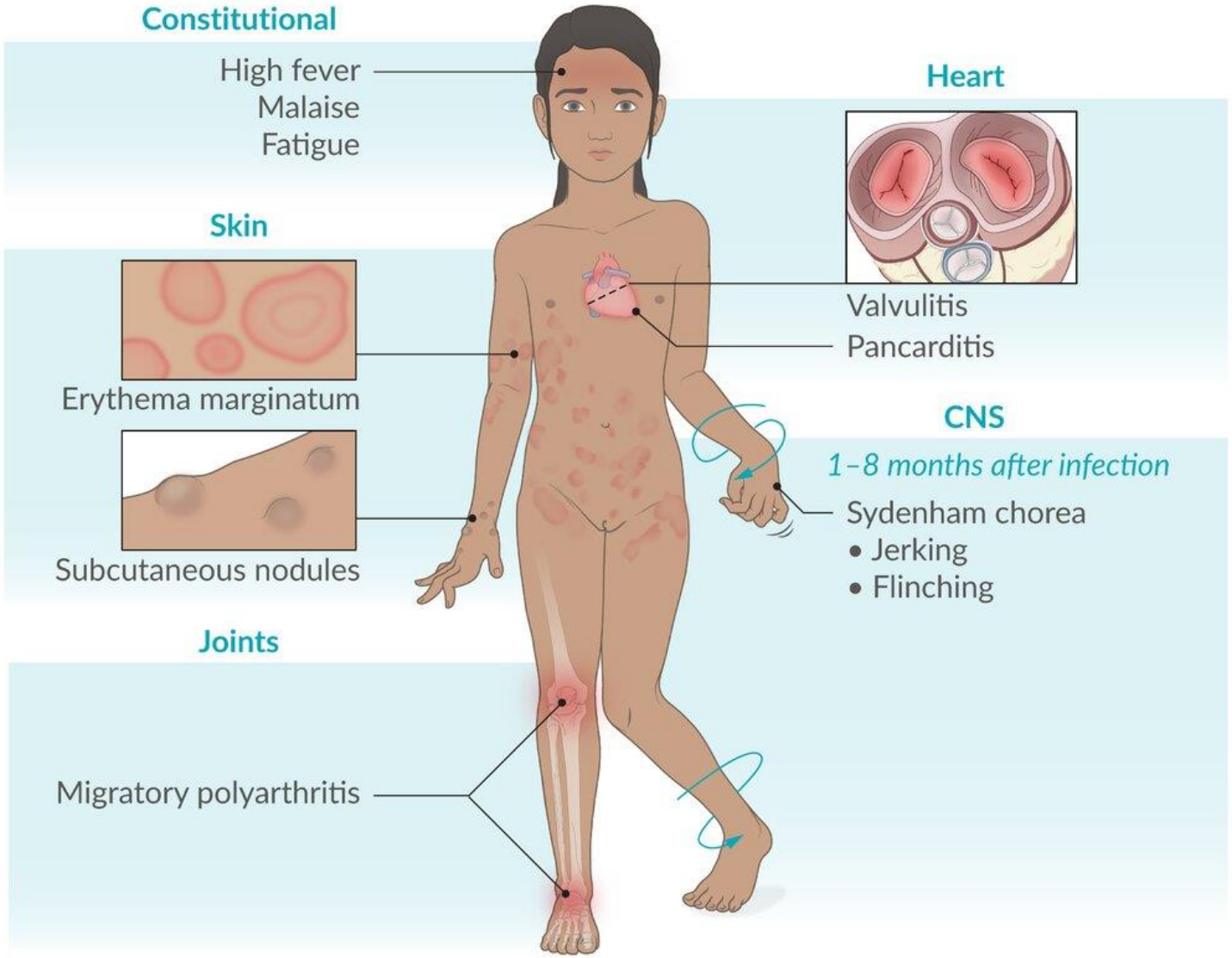
Acute rheumatic fever

Etiology
Previous GAS pharyngitis/tonsillitis without antibiotic treatment

Peak incidence
5–15 years of age

Diagnosis
Evidence of a previous GAS infection (e.g., throat culture, rapid antigen detection test, antistreptolysin O, antistreptococcal DNase B test)
Revised Jones criteria

Treatment
Antibiotic treatment of underlying GAS infection
Symptomatic treatment of arthritis and fever



Pathophysiology and disease course

Untreated GAS pharyngitis/tonsillitis	Development of antibodies	Molecular mimicry	Type II hypersensitivity reaction	Inflammation and cell damage
Acute infection		Latent period (2–4 weeks)		Rheumatic fever onset

Endemic Diseases

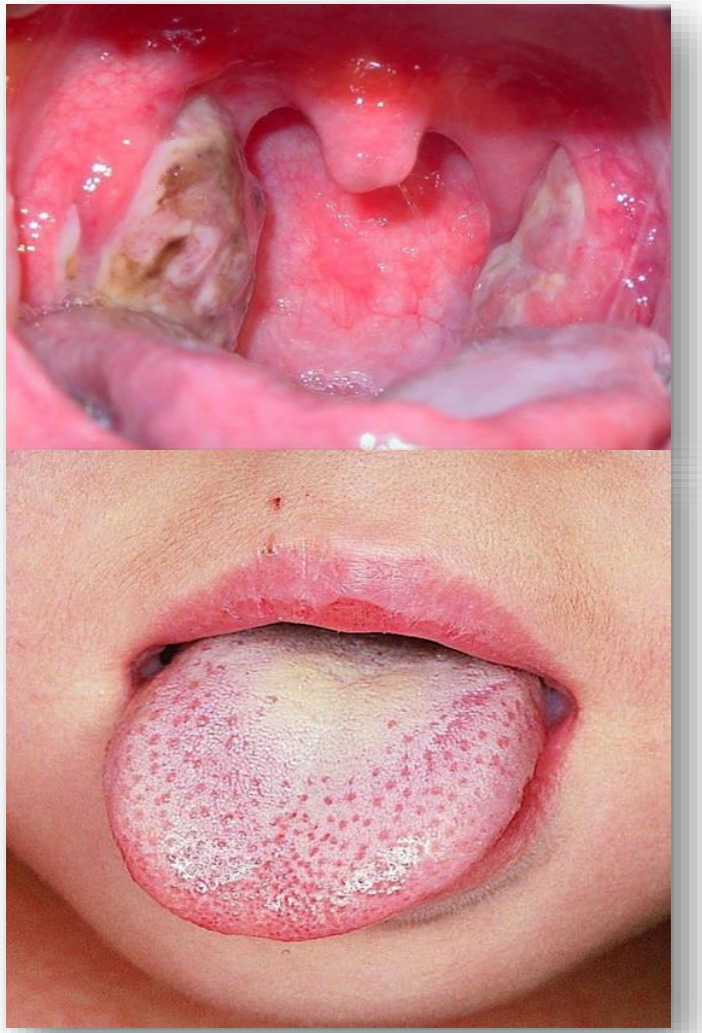
Low-Yield

Infectious mononucleosis

- **Causative agent**: Epstein-Barr Virus (EBV)
- **Route of transmission**: Saliva or respiratory secretions
- **Clinical manifestations**: fever, hepatosplenomegaly, pharyngitis with tonsillar exudates and lymphadenopathy
- **Diagnostics**:
  - **Monospot test**
  - **Serology**: Anti-viral capsid antigen antibodies (anti-VCA), Anti-EBV nuclear antigen-antibody (anti-EBNA-1)
  - **PCR**
  - **Peripheral blood smear**: lymphocytosis with > 10% atypical lymphocytes (in some cases, up to 90%)
- **Serious complication**: Hodgkin's lymphoma, Nasopharyngeal carcinoma

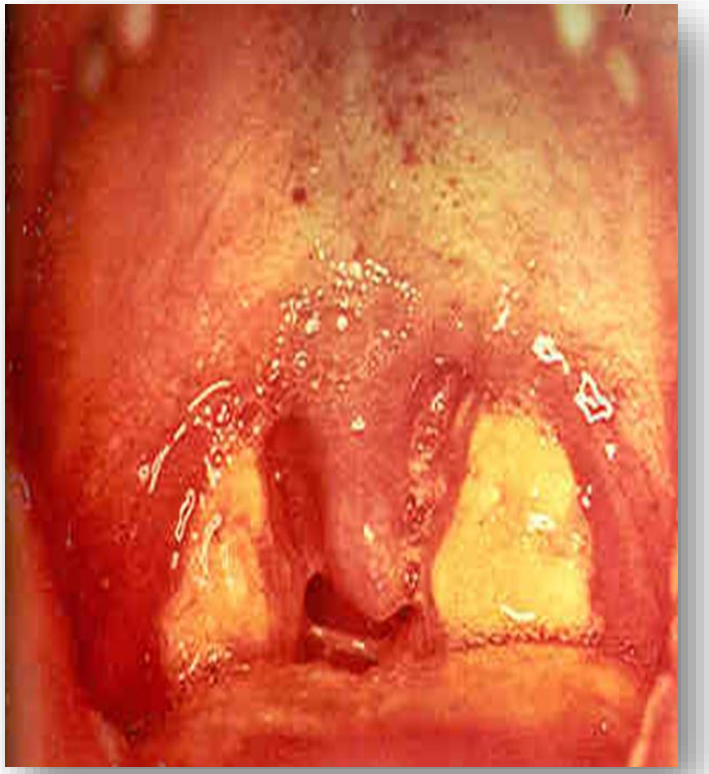
This boy presented with cervical lymphadenopathy + splenomegaly

- **What investigation would you like to ask for?**
  - Diagnosed by Mono spot test, Serology (anti-VCA, anti-EBNA-1), PCR
- **What is your differential diagnosis ?**
  - Infectious mononucleosis



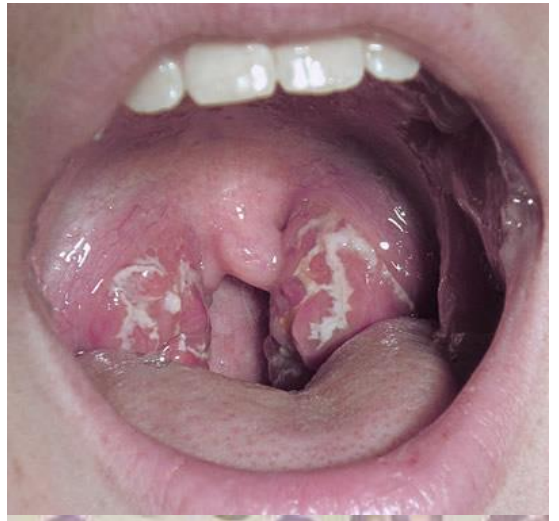
Infectious mononucleosis

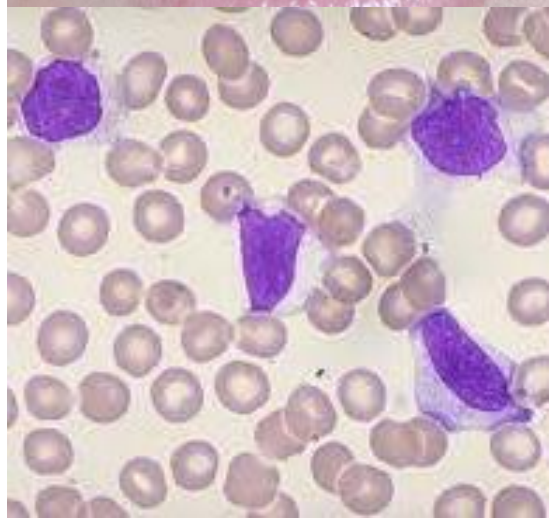
- 3 years old male presented to ED with fever / cough on examination hepatosplenomegaly is found with cervical LAP
- **What's causative organism ?**
  - EBV
- **What's the complication ?**
  - Spleen rupture
- **What investigation would you like to ask for?**
  - Diagnosed by Mono spot test, Serology (anti-VCA, anti-EBNA-1), PCR



Infectious mononucleosis

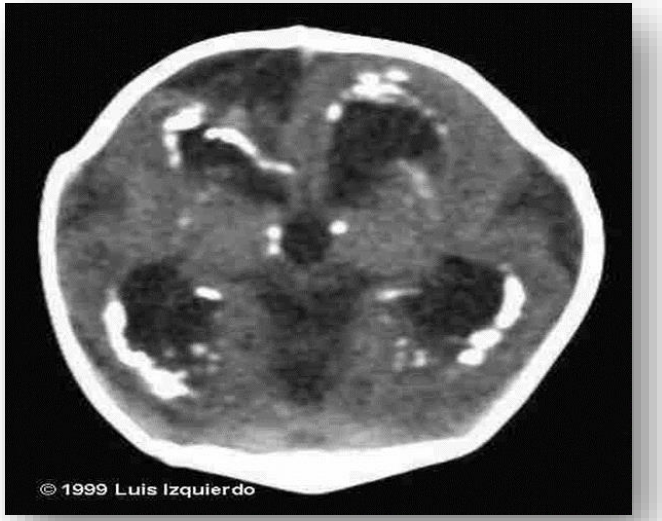
- **What is your differential diagnosis ?**
  - Infectious mononucleosis
- **Describe what you see in this blood film ?**
  - Atypical lymphocytes
- **Most common organism ?**
  - EBV
- **If this patient develop rash after give him antibiotics. What type of this antibiotics ?**
  - Penicillins especially Amoxicillin or Ampicillin



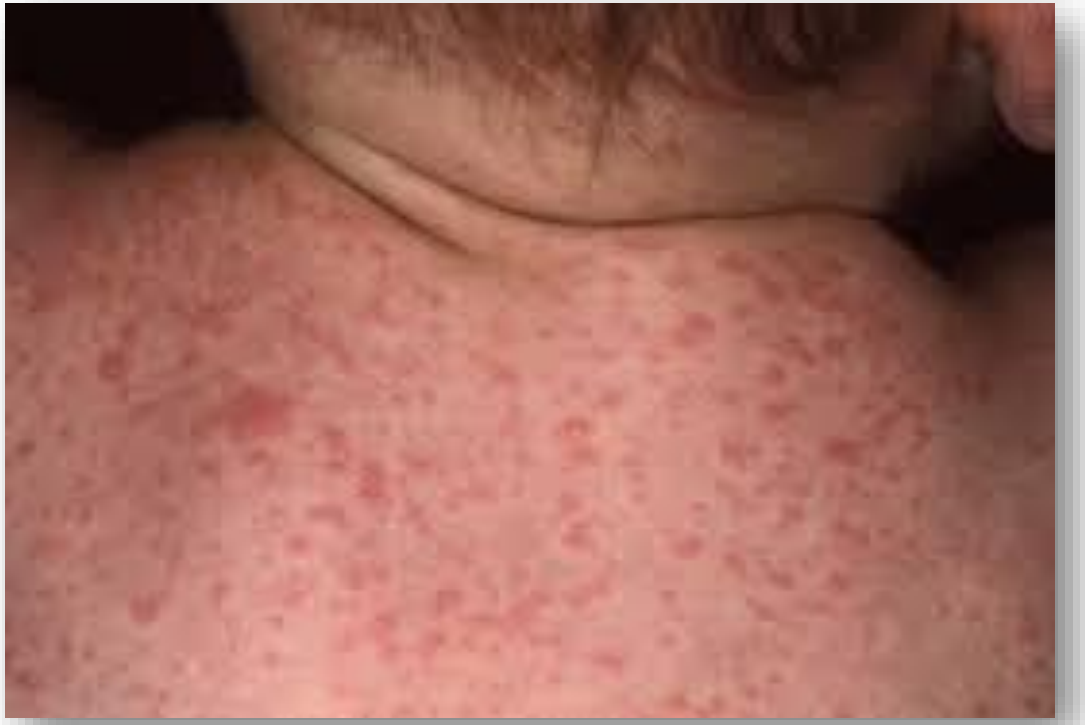


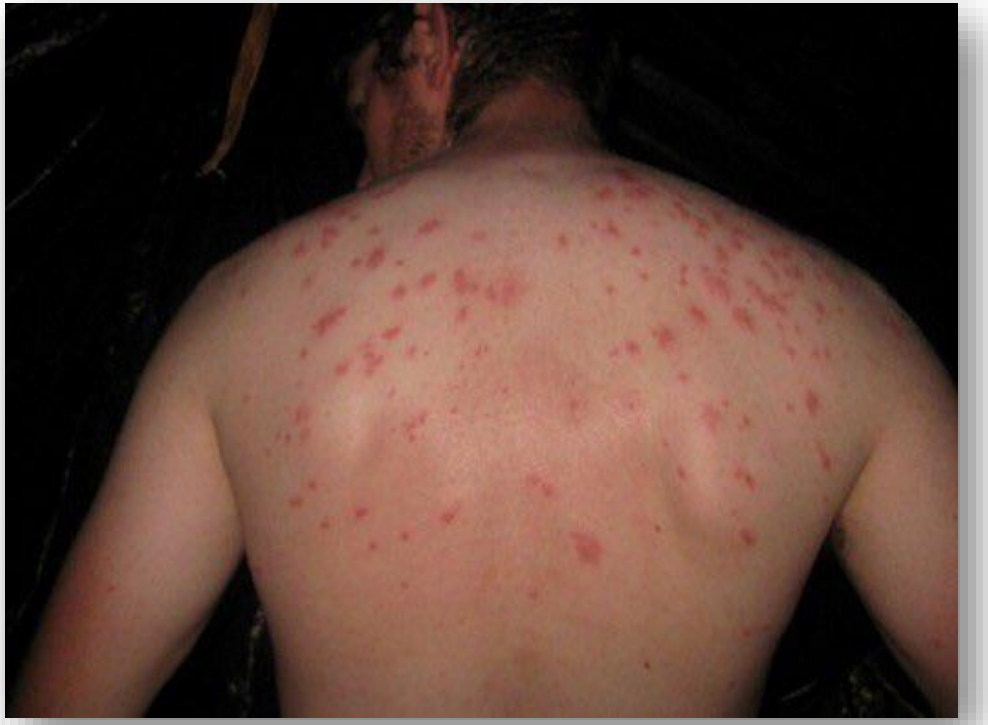
Infectious mononucleosis

- **What's the most probable diagnosis ?**
  - Peri-Ventricular calcifications
- **Describe what you see**
  - congenital CMV



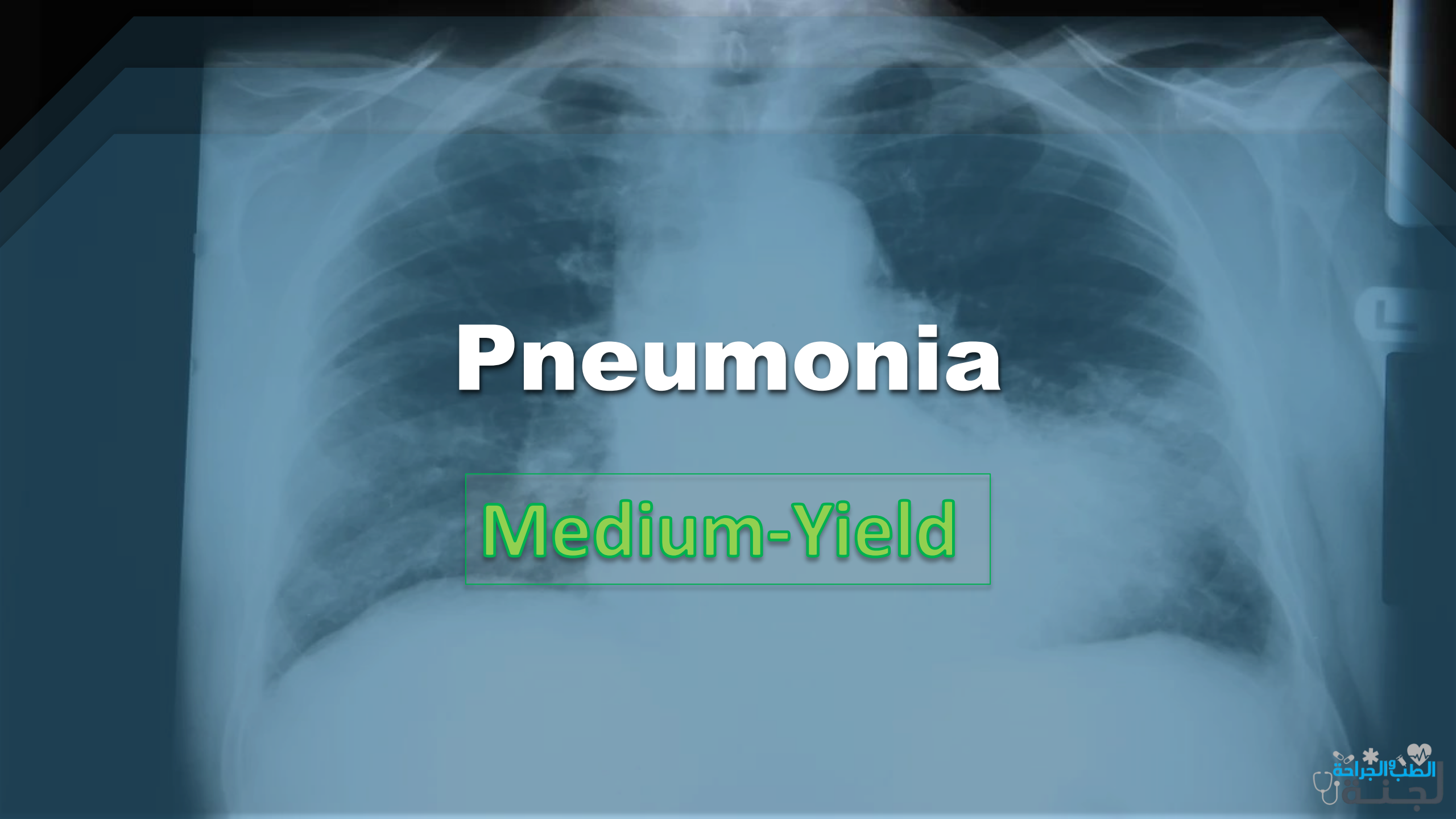
Typhoid and paratyphoid fever





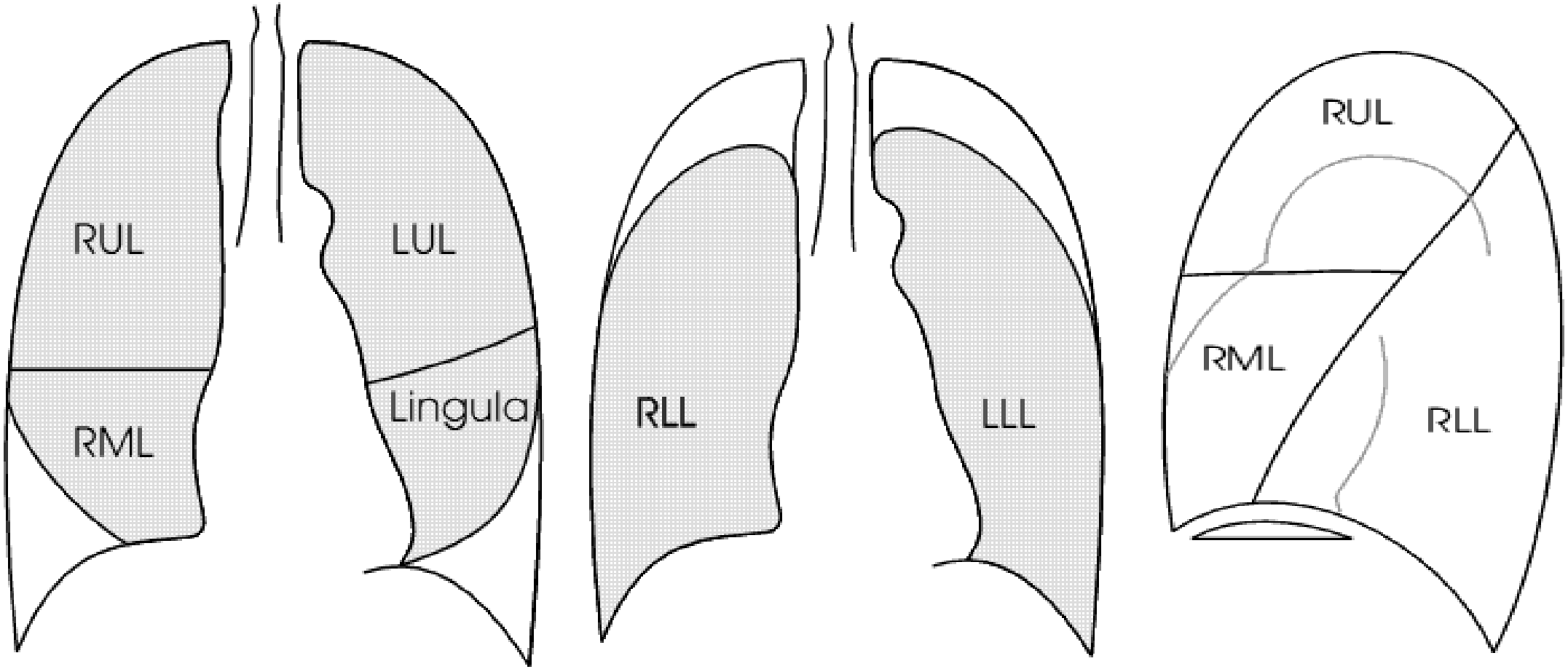
- **Rose-colored spots**: a small, speckled, rose-colored exanthem that appears on the lower chest and abdomen (most commonly around the navel) in approx. 30% of affected individuals

Respiratory

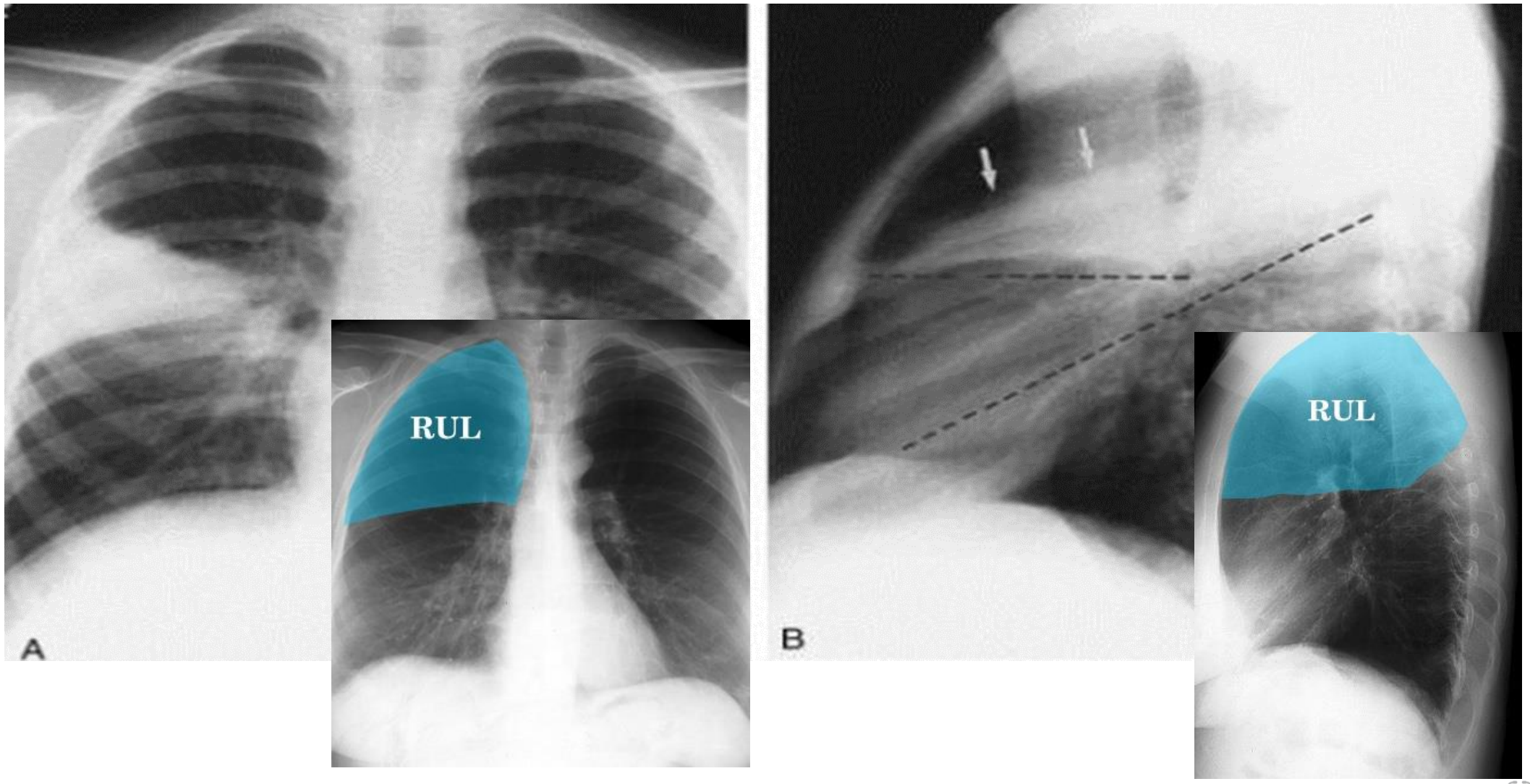

Pneumonia
Medium-Yield

إضافي

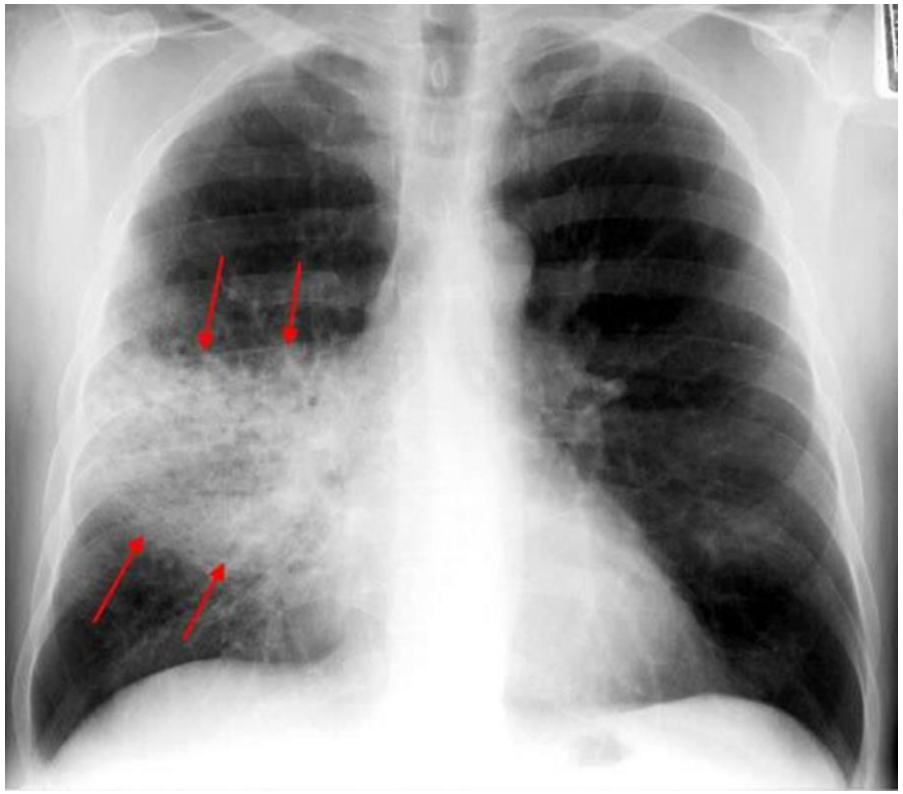
Lung Anatomy on Chest X-ray

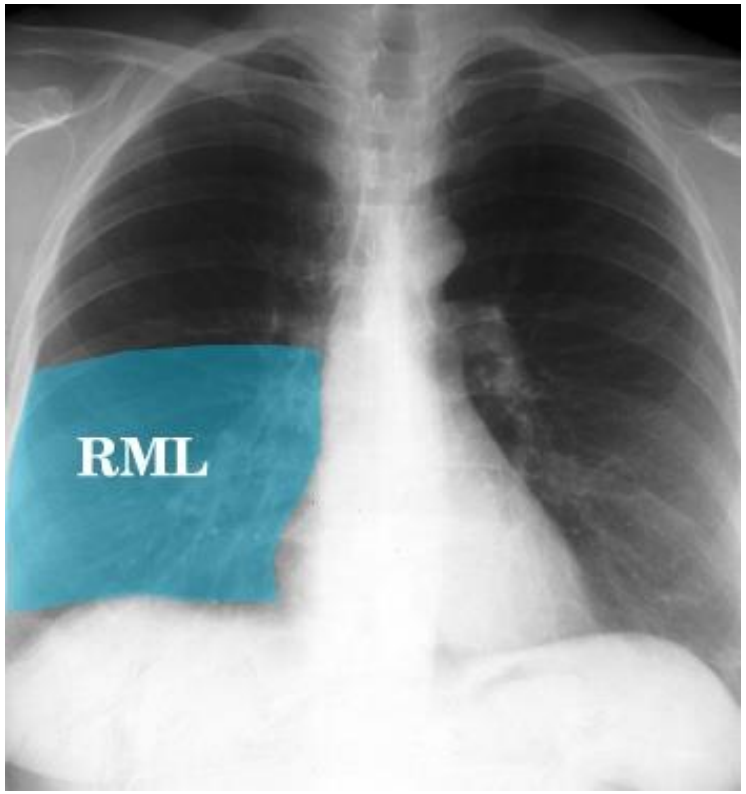


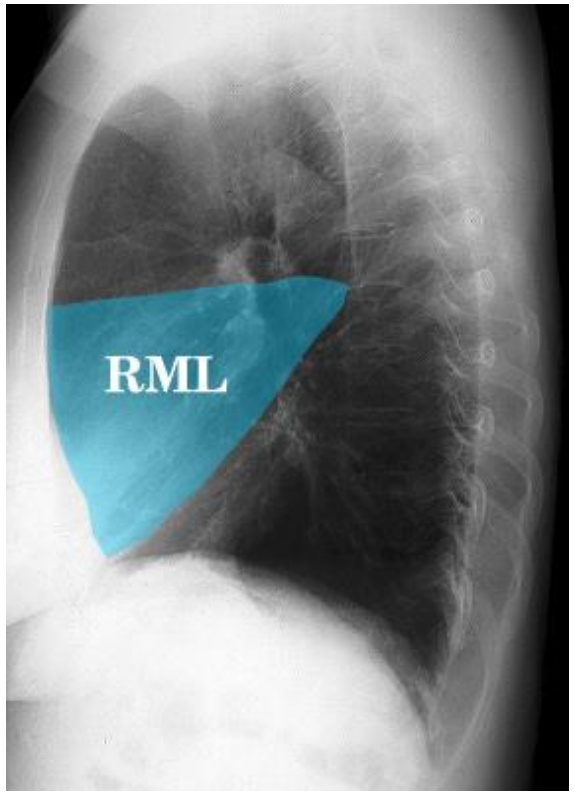
Right upper lobe pneumonia



Right middle lobe pneumonia







Right lower lobe pneumonia

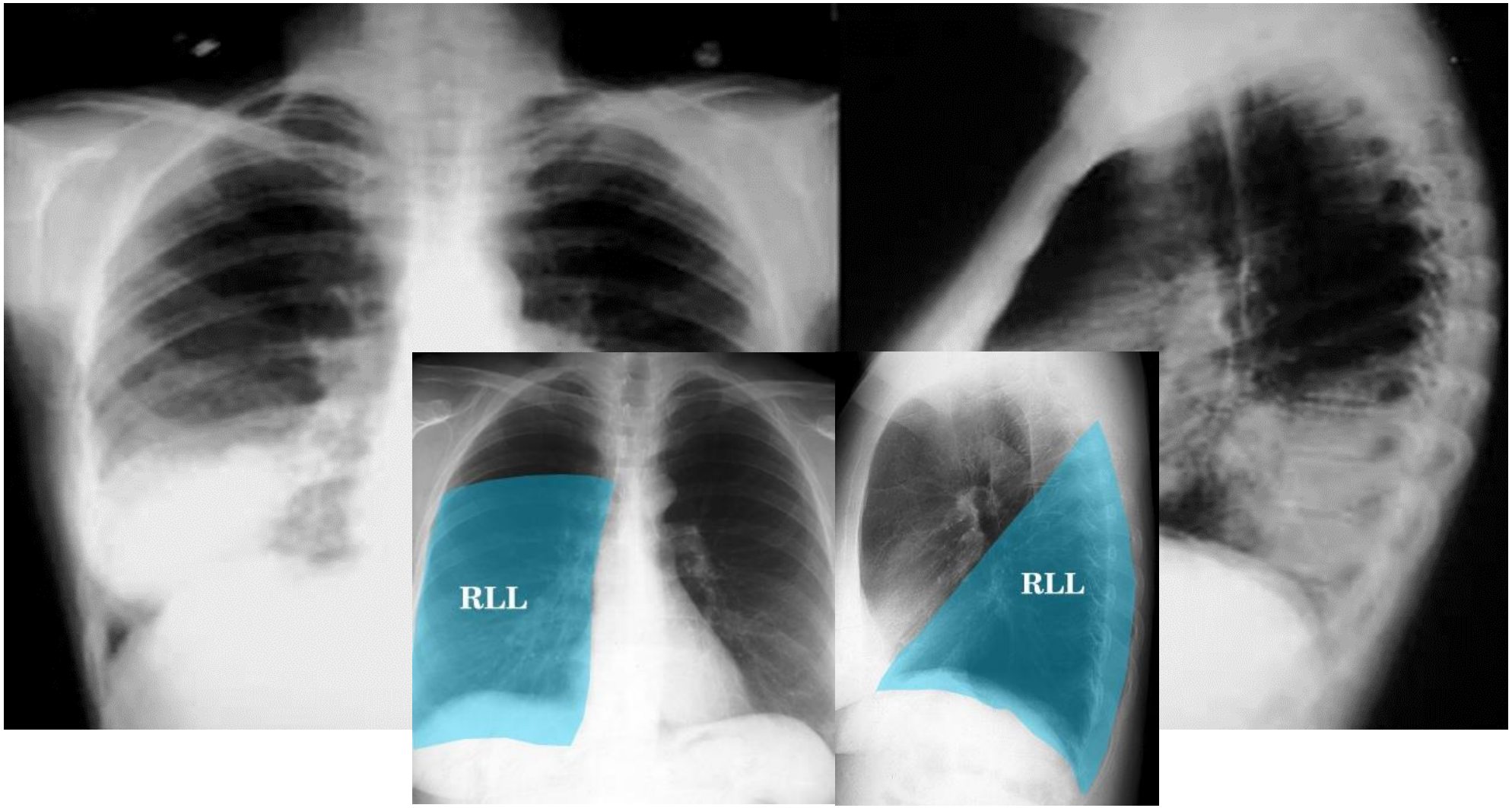


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Table 25.1 Most Common Agents Causing Community-Acquired Pneumonia According to Age Group

AGE				
Newborns	**1–6 Months**	**6–12 Months**	**1–5 Years**	**Older Than 5 Years**
Group B Streptococcus Enteric Gram-negative RSV	Viruses *Streptococcus pneumoniae* *Haemophilus influenzae* *Staphylococcus aureus* *Moraxella catarrhalis* *Chlamydia trachomatis* *Ureaplasma urealyticum* *Bordetella pertussis*	Viruses *Streptococcus pneumoniae* *Haemophilus influenza* *S. aureus* *Moraxella catarrhalis*	Viruses *M. pneumoniae* *S. pneumoniae* *C. pneumoniae*	Viruses *M. pneumoniae* *S. pneumoniae* *C. pneumoniae*

Table 25.2 Choice of Antibiotic Treatment for Community-Acquired Pneumonia When Typical Bacteria Are Identified

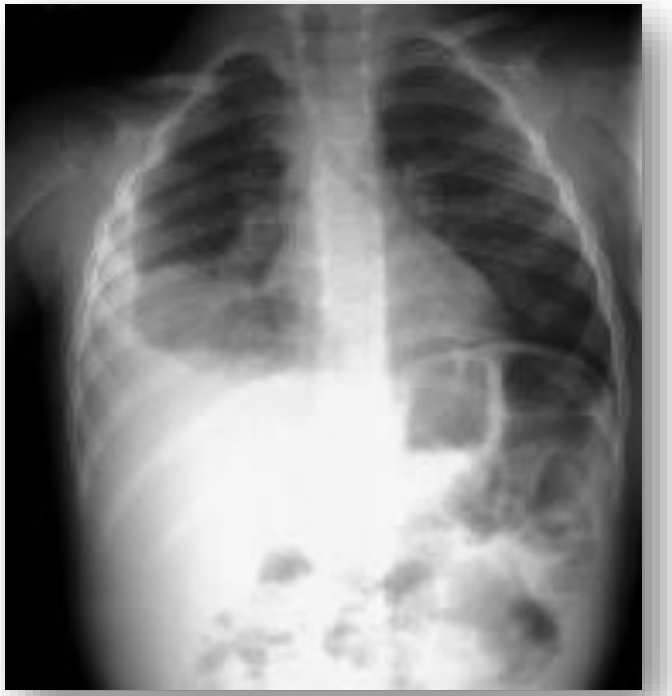
Pathogen	First Choice	Other
Streptococcus pneumoniae, penicillin susceptible or intermediate	Penicillin, ampicillin, or high-dose amoxicillin	Cefuroxime, ceftriaxone, azithromycin
S. pneumoniae, penicillin resistant (MIC ≥ 4 µg/mL)	Second- or third-generation cephalosporins for sensitive strains; vancomycin	
Staphylococcus aureus	Methicillin/oxacillin	Vancomycin or teicoplanin (for MRSA)
Haemophilus influenzae	Amoxicillin	Amoxicillin/clavulanate, cefuroxime, ceftriaxone, other second- and third-generation cephalosporins
Moraxella catarrhalis	Amoxicillin/clavulanate	Cefuroxime

Treatment

- **First line treatment in CAP**: oral amoxicillin (high dose) or IV ampicillin if IV treatment is required
- Macrolide should be added when pneumonia due atypical bacteria is suspected
- Vancomycin or teicoplanin should be reserved for severely ill patients, when coverage for highly resistant pneumococcus is desired
- **Neonates with CAP**: IV ampicillin and gentamicin
- Whenever there is a positive culture or a clinical picture suggestive of S. aureus, specific antibiotic coverage against this pathogen should be added (e.g., methicillin, oxacillin, clindamycin, or vancomycin in the case of MRSA strains).

Pneumonia

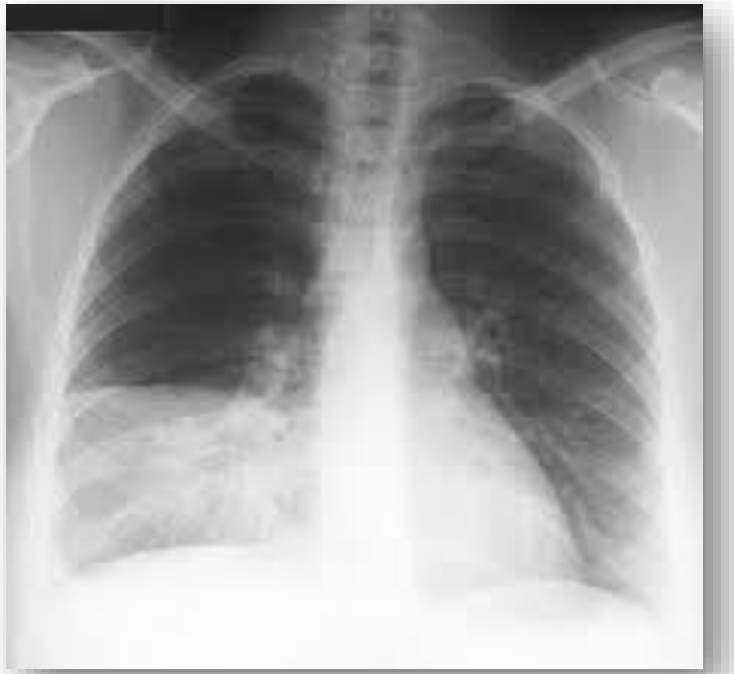
- 7-year-old male complained of SOB, cough & fever, the vitals: hypotension, the patient look sick, CXR is shown.



- **What are the abnormalities in this x-ray film ?**
  - Area of consolidation in the right lower lobe with massive pleural effusion (meniscal sign + visible cardiac silhouette indicates the consolidation is in the lower not middle lobe)
- **What is the treatment ?**
  - Oral amoxicillin high dose (1 g PO every 8 hours)
- **Write 2 complications ?**
  - Parapneumonic pleuritis, Parapneumonic pleural effusion, Pleural empyema, Lung abscess, ARDS, Respiratory failure, Sepsis

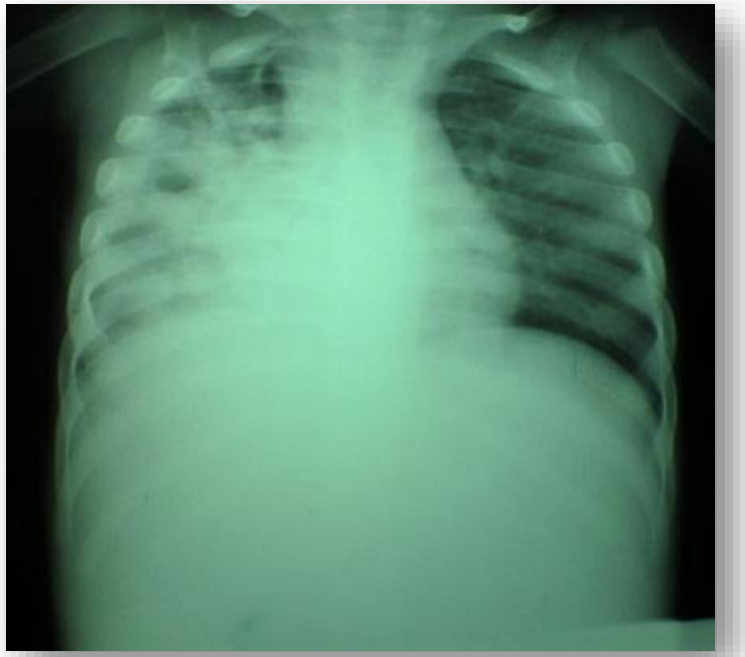
Pneumonia

- **What is the findings in this Xray ?**
  - Middle lobe pneumonia (lobar pneumonia)
- **What Is your choice of treatment ?**
  - Oral amoxicillin high dose (1 g PO every 8 hours)



Pneumonia

- 6-year-old boy came to your clinic complaining of cough and fever



- **Mention the prominent finding**
  - Heterogenous opacification on the right lung field
- **What is the most likely Diagnosis ?**
  - Right interstitial pneumonia (atypical pneumonia)
- **What the most common microorganism ?**
  - Mycoplasma pneumoniae
  - Chlamydia pneumoniae
- **Treatment**: Macrolides

Pneumonia

- Fever and cough, RR =33

❖ **What are 2 physical finding on chest exam without using stethoscope?**

- Dullness in percussion in Rt. Side.
- Asymmetrical chest expansion.

❖ **What are 2 findings in CXR?**

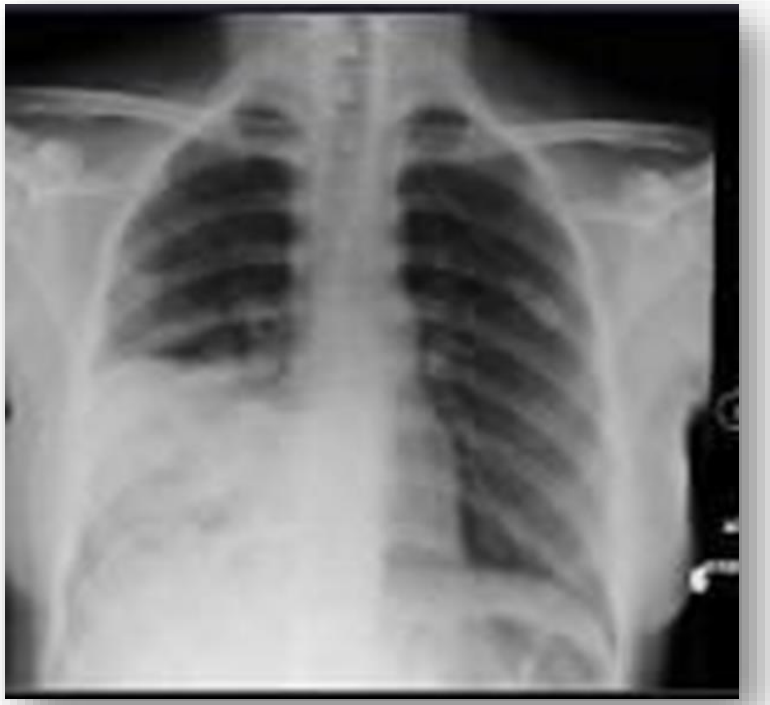
- Hyper-dense or consolidation on Rt. side.
- Costo-phrenic angle obliterated or absent.

❖ **What's the most likely Dx.?**

- Rt. Lower and middle lobe Pneumonia.

❖ **What's the most common microorganism?**

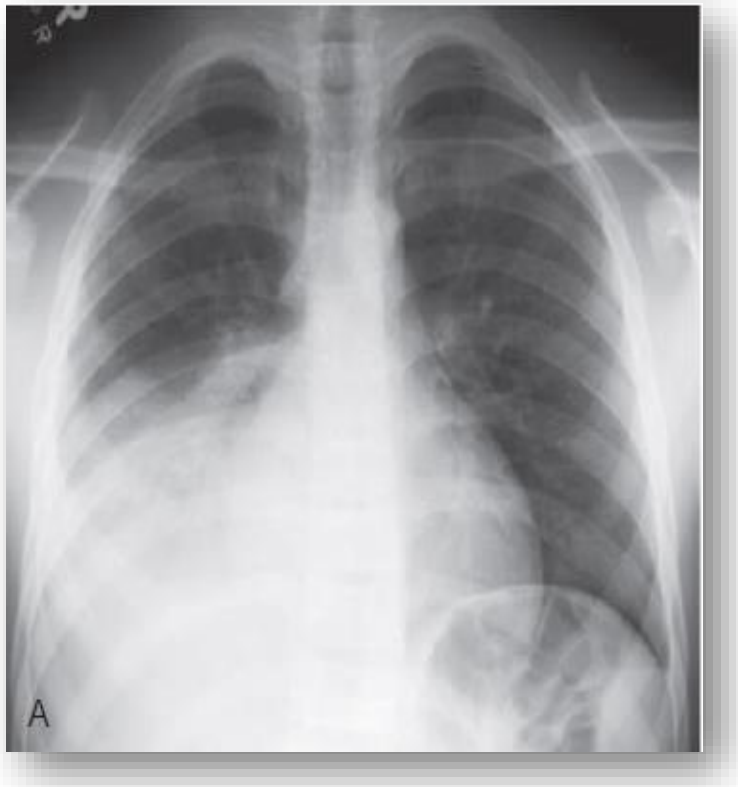
- Strep. Pneumonia.



CURB-65 score = 1
No need for admission

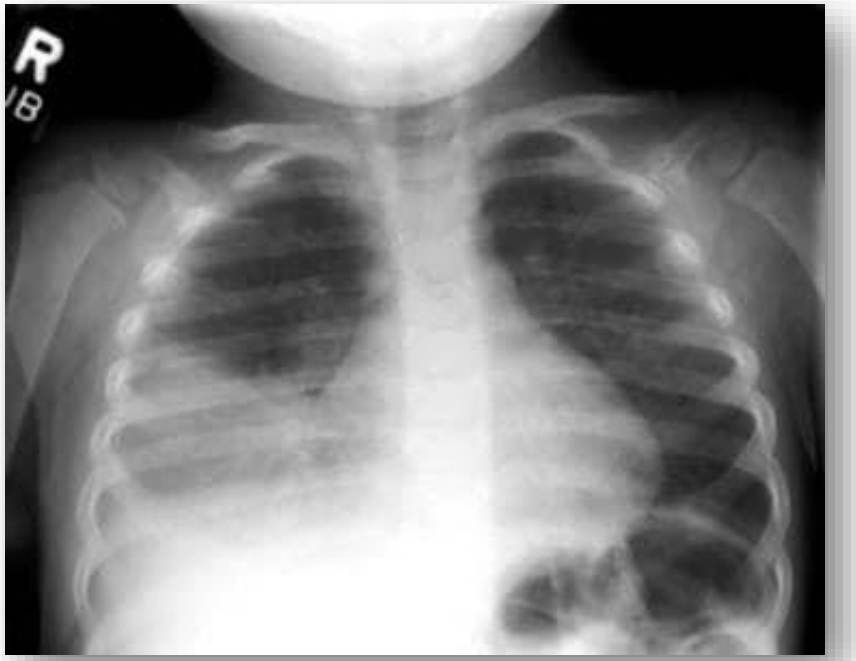
Pneumonia

- 6 years old patient presented with this CXR
- **What is the most common organism causing this finding ?**
  - S.pneumonae
- **What is your management ?**
  - Oral amoxicillin high dose (1 g PO every 8 hours)



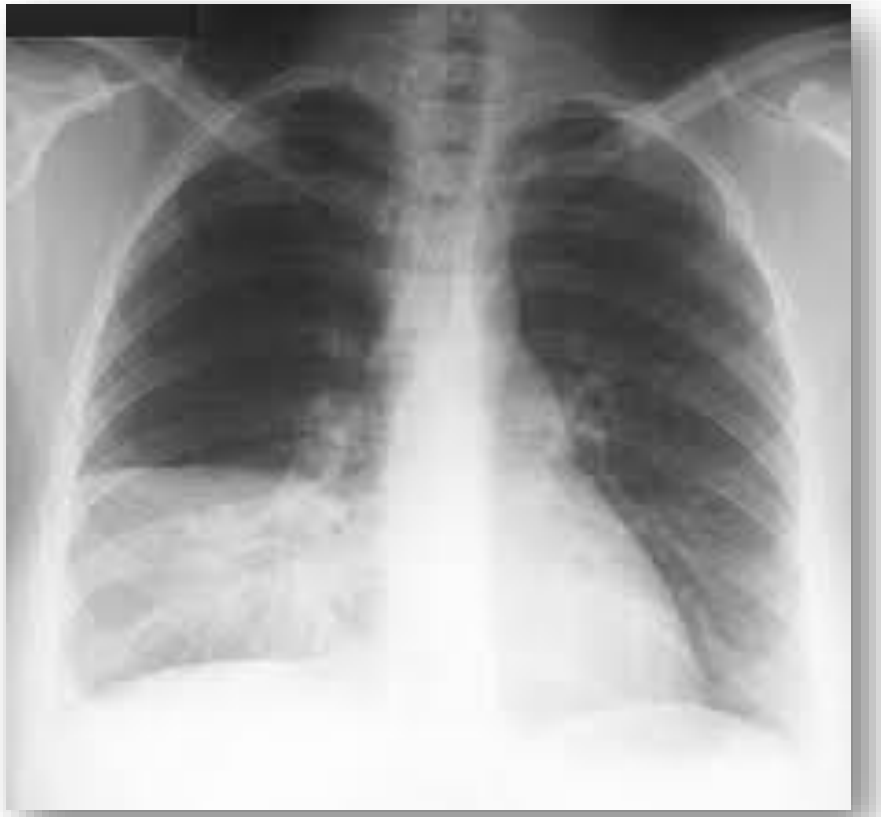
Pneumonia

- **Describe this chest x ray**
  - Right lower & middle lobe consolidation (silhouetting with diaphragm and the heart and obliteration of right costophrenic angel)
- **Most common organism**
  - Streptococcus pneumoniae
- **Drug of choice**
  - Oral amoxicillin high dose (1 g PO every 8 hours)



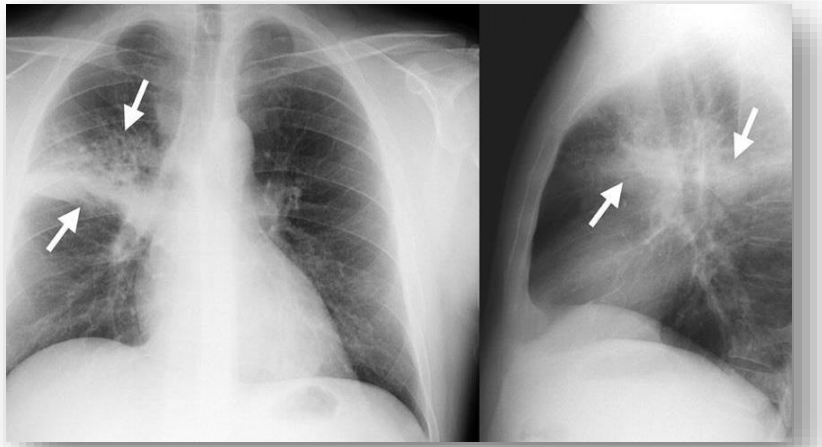
Pneumonia

- **Describe this chest x ray**
  - Right middle lobe consolidation (silhouetting with diaphragm and the heart and obliteration of right costophrenic angel)
- **Drug of choice**
  - Oral amoxicillin high dose (1 g PO every 8 hours)



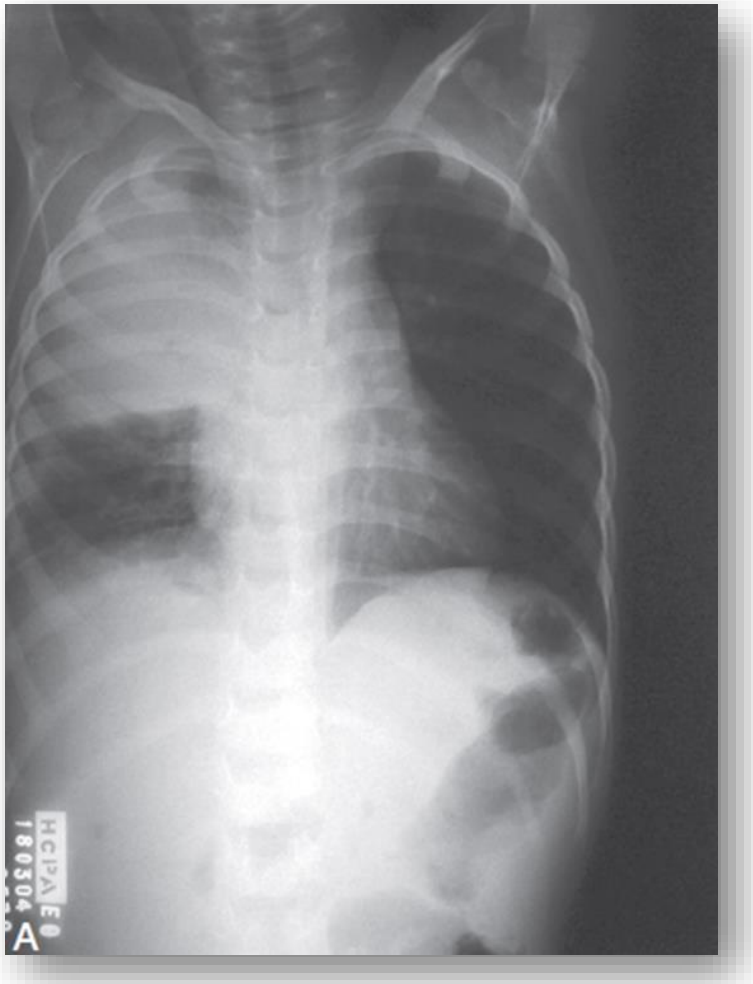
Pneumonia – Cystic fibrosis

- 10-year-old patient who had recurrent chest infections, and FTT
- **How to confirm your diagnosis ?**
  - Chloride sweat test >60 on 2 separated days or genetic studies (more than 2 mutations)
- **Mention two microorganisms could cause this picture at his age ?**
  - S.aureus (common at this age)
  - Pseudomonas aeruginosa (common at this age)
  - H.infleunzae (common at this age)
  - Burkholderia cepacia (rare)



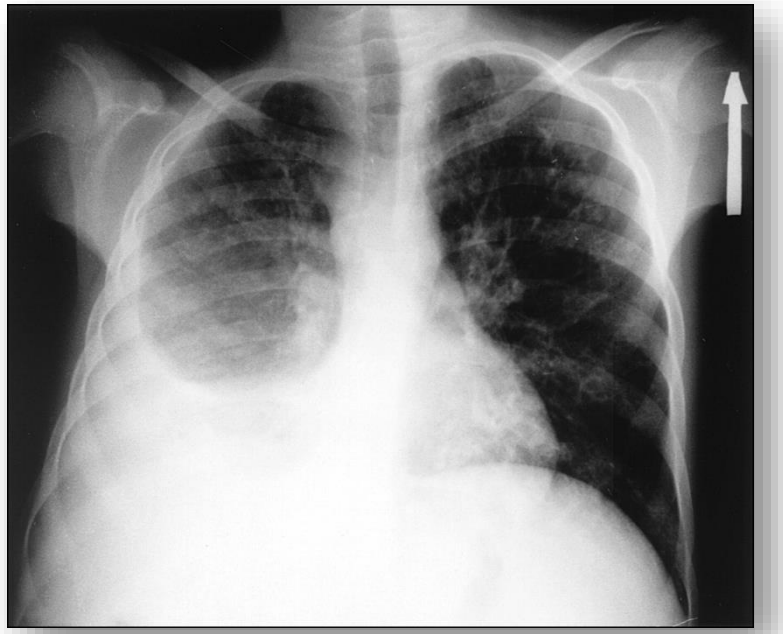
Pneumonia

- 4 y old girl come to ED with high grade fever and sever cough and low oxygen saturation
- **What's the finding of this X-ray ?**
  - Right upper lobe consolidation
- **Mention 2 DDx**
  - Lobar pneumonia
  - Atelectasis
- **What's the organism?**
  - Pneumococcus
- **What's the management ?**
  - Admission + O_2 + antibiotic + antipyretic



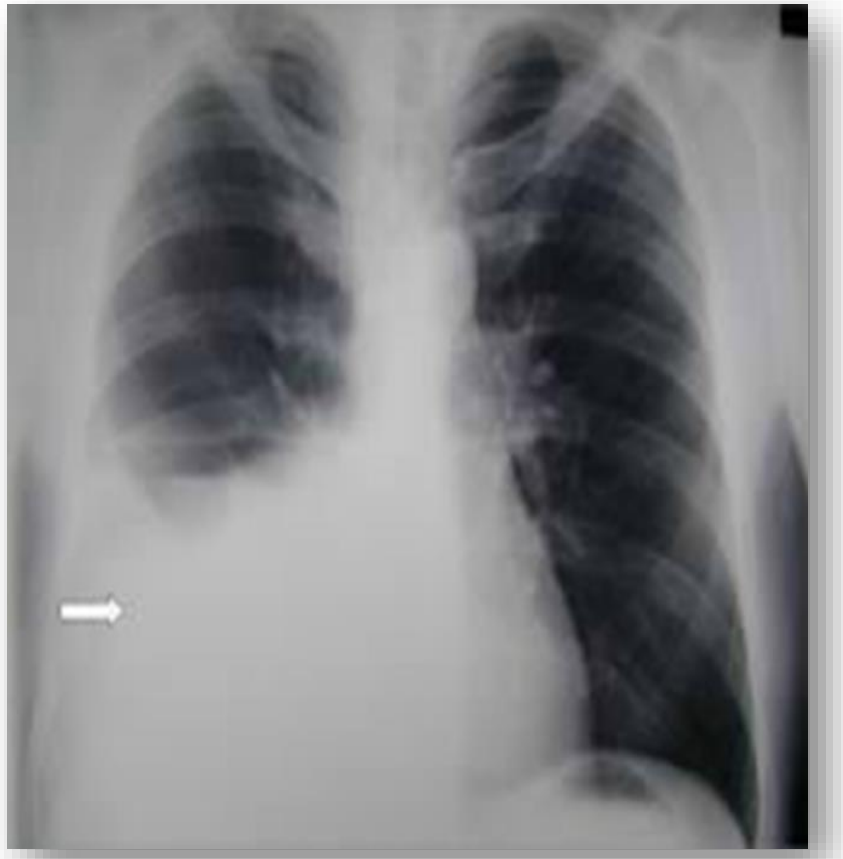
Pneumonia

- Patient present with wet cough, fever
- **What are the chest x-ray findings ?**
  - Right lung homogenous opacification
  - Silhouetting with heart and diaphragm
  - Meniscus sign
- **Mention 2 DDx**
  - Pneumonia, Pleural effusion
- **What is your next imaging to confirm Dx ?**
  - CT scan
- **What is your management ?**
  - Oral amoxicillin high dose (1 g PO every 8 hours)



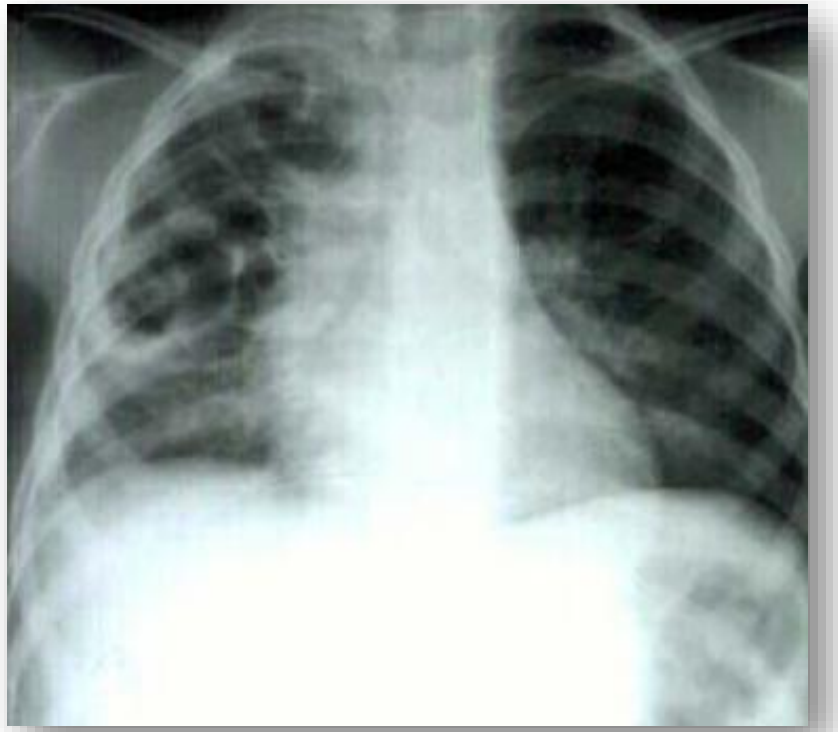
Pneumonia

- **What's your Dx ?**
  - Rt. sided pleural effusion.
- **Give 2 findings in the chest exam (not by auscultation)**
  - Stony dull percussion.
  - Decreased chest expansion on Rt. Side (on palpation).
- **Give auscultation findings**
  - Diminished vesicular breathing on Rt. Side
  - Bronchial breathing & crackles
- **What's the treatment ?**
  - Chest tube, Antibiotic



4Y/O patient presented with high grade fever, lethargy and wet cough

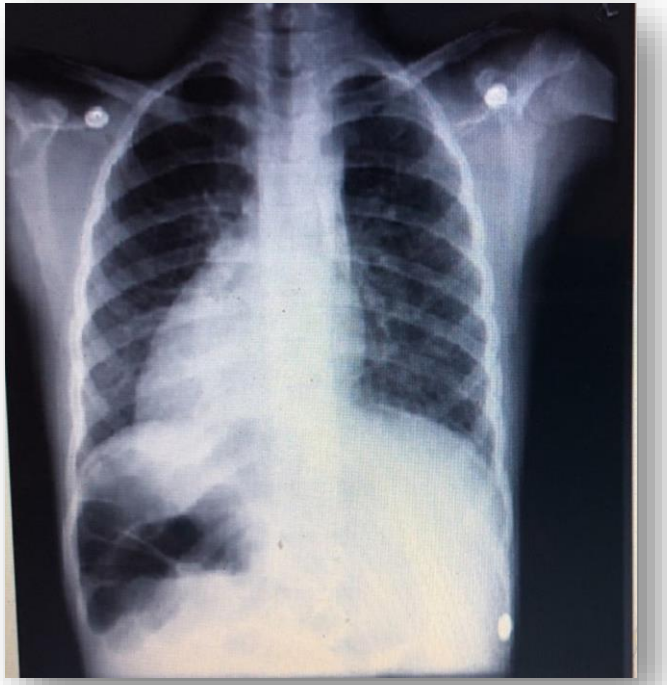
- **What is the diagnosis ?**
  - Pneumatocele
- **What is the cause ?**
  - Staph aureus
- **What is the gold standard diagnosis ?**
  - Culture
- **What is the best treatment ?**
  - Antibiotic covering S.aureus (e.g., methicillin) to treat the underlying infection
  - Supportive and includes monitoring and observation, may require surgical management
- **Another DDx**
  - Lung abscess, secondary TB



Kartagener syndrome

- This chest x-ray for 6 years old male patient presented with chronic productive cough.

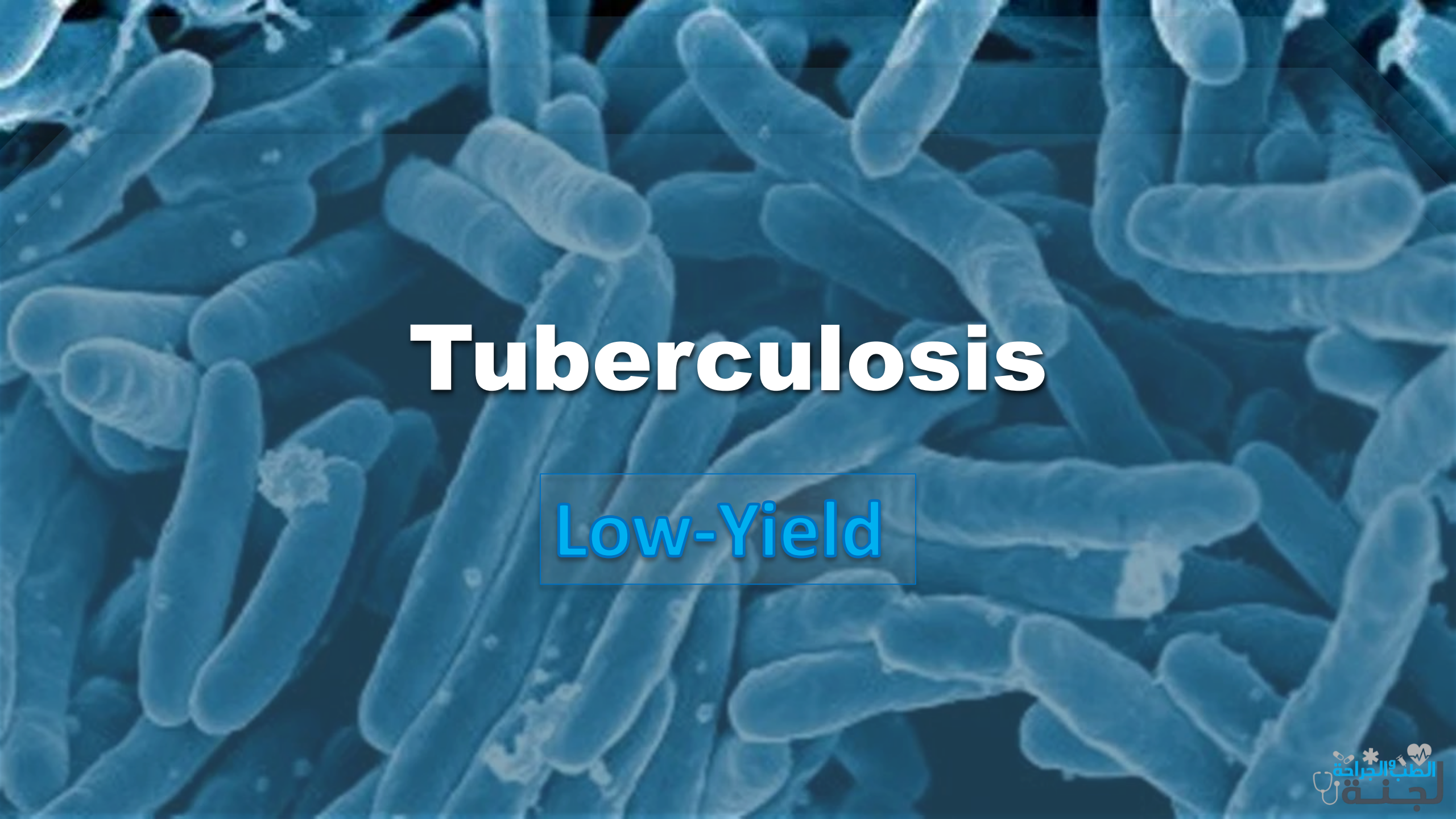
- All of the following are true except:
  a. Shows dextrocardia and situs inversus totalis
  b. Patients with this condition are more likely to have recurrent pneumonia
  c. Failure to thrive is an early manifestation of this condition
  d. Recurrent otitis media and sinusitis can occur in this condition
  e. Bronchiectasis is a long-term complication



Kartagener syndrome classic triad: situs inversus, recurrent sinusitis, and bronchiectasis

TABLE **110.1** Etiologic Agents and Empirical Antimicrobial Therapy for Pneumonia in Patients Without History of Recent Antibiotic Therapy

AGE GROUP	COMMON PATHOGENS* (IN APPROXIMATE ORDER OF FREQUENCY)*	LESS COMMON PATHOGENS	OUTPATIENTS (7–10 DAYS TOTAL DURATION OF TREATMENT)†	PATIENTS REQUIRING HOSPITALIZATION (10–14 DAYS TOTAL DURATION OF TREATMENT)‡	PATIENTS REQUIRING INTENSIVE CARE (10–14 DAYS TOTAL DURATION OF TREATMENT)*,‡
Neonates (up to 1 mo of age)	Group B streptococcus, *Escherichia coli*, other gram-negative bacilli, *Streptococcus pneumoniae*	Cytomegalovirus, herpes simplex virus, *Listeria monocytogenes*, *Treponema pallidum*, *Haemophilus influenzae* (type b,§ nontypable)	Outpatient management not recommended	Ampicillin *plus* ceftazidime or an aminoglycoside *plus* an antistaphylococcal agent if *Staphylococcus aureus* is suspected	Ampicillin *plus* ceftazidime or an aminoglycoside *plus* an antistaphylococcal agent if *S. aureus* is suspected
1–3 mo					
Febrile pneumonia	Respiratory syncytial virus, other respiratory viruses (parainfluenza viruses, influenza viruses, adenoviruses), *S. pneumoniae*, *H. influenzae* (type b,§ nontypable)		Initial outpatient management not recommended	Amoxicillin or ampicillin if fully immunized for age for *S. pneumoniae* and *H. influenzae* type b. Alternatives: ceftriaxone if not fully immunized or local *S. pneumoniae* penicillin resistance is significant, with clindamycin if MRSA suspected	Ceftriaxone *plus* nafcillin, oxacillin, clindamycin, or vancomycin
Afebrile pneumonia	*Chlamydia trachomatis*, *Mycoplasma hominis*, *Ureaplasma urealyticum*, cytomegalovirus *Bordetella pertussis*		Erythromycin, azithromycin, or clarithromycin with close follow-up	Erythromycin, azithromycin, or clarithromycin	Erythromycin, azithromycin, or clarithromycin *plus* ceftriaxone *plus* nafcillin, oxacillin, clindamycin, or vancomycin
3 mo to 5 yr	Respiratory syncytial virus, other respiratory viruses (parainfluenza viruses, influenza viruses, human metapneumovirus adenoviruses), *S. pneumoniae*, *H. influenzae* (type b,§ nontypable)	*C. trachomatis*, *Mycoplasma pneumoniae*, *Chlamydophila pneumoniae*, group A streptococcus, *S. aureus*, *Mycobacterium tuberculosis*	Amoxicillin *plus* erythromycin, azithromycin, or clarithromycin if atypical pneumonia suspected	Ampicillin Alternatives: ceftriaxone if not fully immunized or local *S. pneumoniae* penicillin resistance is significant, with clindamycin if MRSA suspected; add erythromycin, azithromycin, or clarithromycin if atypical pneumonia suspected	Cefuroxime or ceftriaxone *plus* azithromycin, erythromycin, or clarithromycin *with or without* clindamycin or vancomycin
5–18 yr	*M. pneumoniae*, *S. pneumoniae*, *C. pneumonia*	*H. influenzae* (type b,§ nontypable), influenza viruses, adenoviruses, coronaviruses, other respiratory viruses	Amoxicillin *plus* erythromycin, azithromycin, or clarithromycin if atypical pneumonia suspected	Ampicillin *plus* erythromycin, azithromycin, or clarithromycin if atypical pneumonia suspected	Cefuroxime or ceftriaxone *plus* azithromycin, erythromycin, or clarithromycin *with or without* clindamycin or vancomycin
≥18 yr§	*M. pneumoniae*, *S. pneumoniae*, *C. pneumoniae*, *H. influenzae* (type b,§ nontypable), influenza and coronaviruses, adenoviruses	*Legionella pneumophila*, *M. tuberculosis*	Amoxicillin, *or* erythromycin, azithromycin, clarithromycin, doxycycline, moxifloxacin, gatifloxacin, levofloxacin, or gemifloxacin‖ if atypical pneumonia suspected	Ampicillin *plus* erythromycin, azithromycin, or clarithromycin if atypical pneumonia suspected or moxifloxacin, gatifloxacin, levofloxacin, or gemifloxacin	Ceftriaxone, *plus either* azithromycin or clarithromycin *with or without* clindamycin or vancomycin, or moxifloxacin, gatifloxacin, levofloxacin, or gemifloxacin *with or without* clindamycin or vancomycin


Tuberculosis
Low-Yield

إضافي

Tuberculosis (TB)

Epidemiology
Sex: ♂ >♀ (2:1)
Latent infection: ¼ of world population
Incidence (USA.): ~ 2.8 per 100,000
One of the leading causes of death worldwide!

Etiology
Mycobacterium tuberculosis complex
(*M. tuberculosis* (95%), *M. bovis*, *M. africanum*)

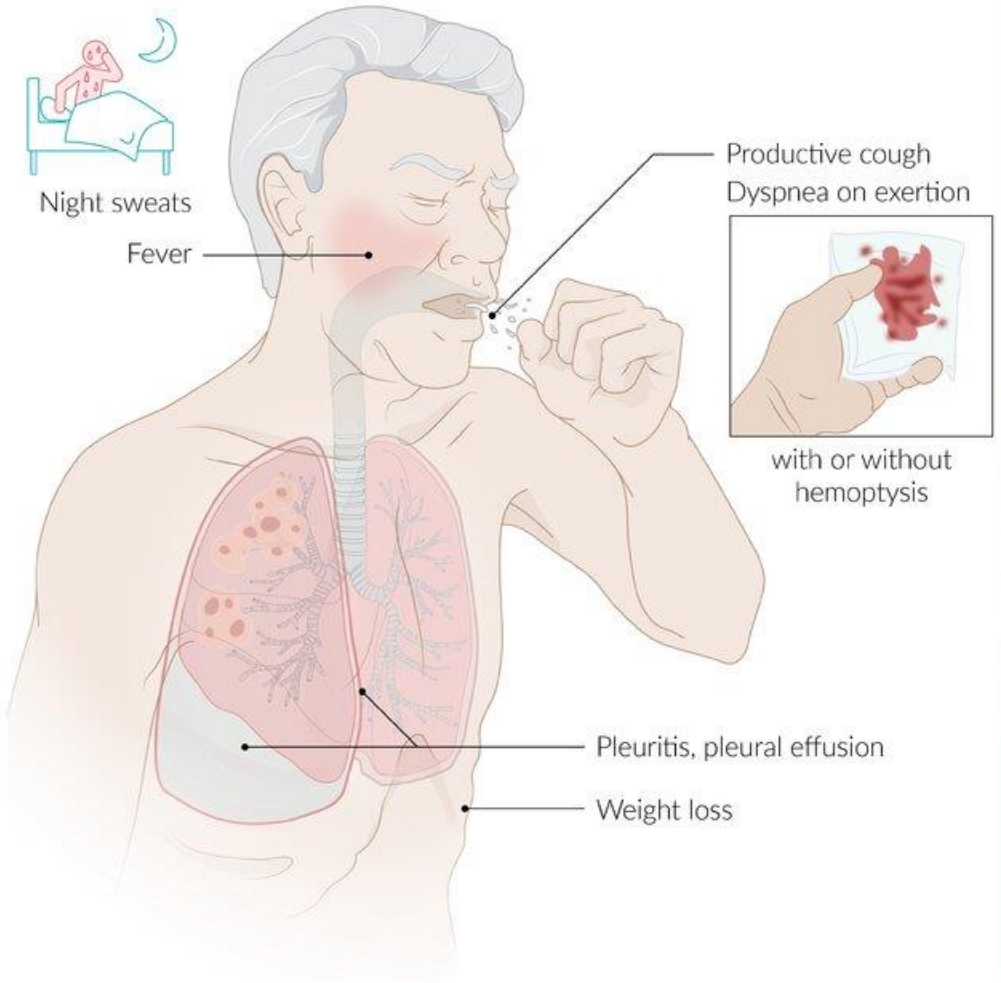
Diagnosis
Chest x-ray: upper lobe nodular or cavitary lesion/s (reactivation TB); ghon complex (primary TB)
Tests for latent TB: tuberculin skin test; interferon-γ release assay
Confirmatory tests: microbiological detection of pathogen (e.g., via PCR, culture and microscopy)

Complications
Miliary tuberculosis, severe tuberculosis sepsis (Landouzy sepsis)

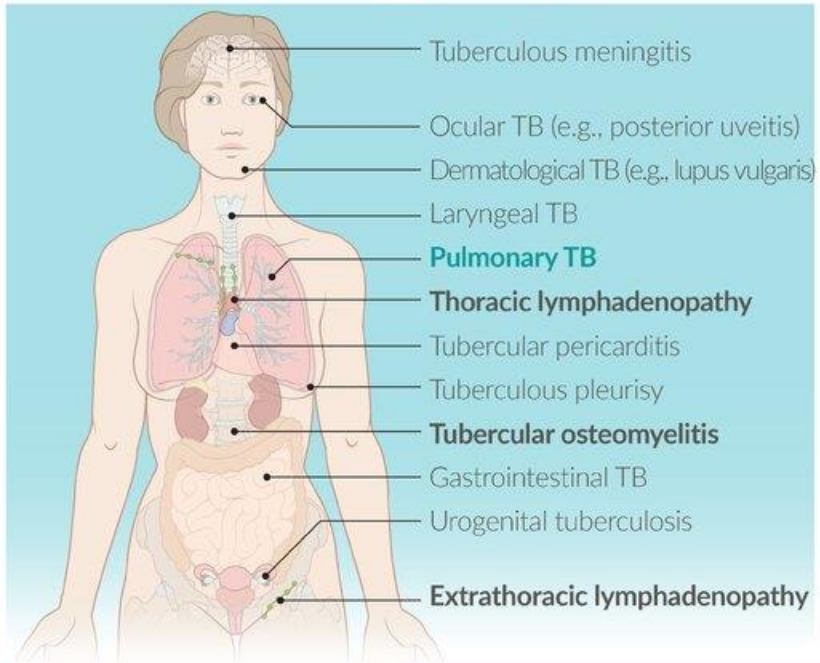
Note
There is a rise of multidrug-resistant tuberculosis infection Long-term combination antibiotic therapy is essential

Risk factors
Immunosuppression
(e.g., HIV, malnutrition, very young or old age), contact with a patient with active TB

Pulmonary TB



Manifestations of TB



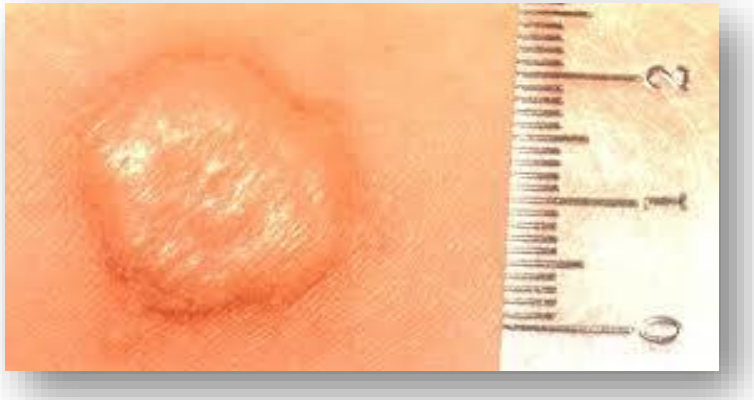
Classification

Primary TB
Latent tuberculosis infection (> 95%): asymptomatic
Active primary tuberculosis (< 5%): often asymptomatic; nonspecific symptoms, such as productive cough, exertional dyspnea, may be present
Reactivation TB (postprimary or secondary TB)
Pulmonary TB (80%)
Extrapulmonary TB (20%)

PPD/TST induration	Patients to treat
≥5 mm	• HIV-positive patients • Recent contacts of known TB case • Nodular or fibrotic changes on chest x-ray consistent with previously healed TB • Organ transplant recipients & other immunosuppressed patients
≥10 mm	• Recent immigrants (<5 years) from TB-endemic areas • Injection drug users • Residents & employees of high-risk settings (eg, prisons, nursing homes, hospitals, homeless shelters) • Mycobacteriology laboratory personnel • Higher risk for TB reactivation (eg, diabetes, prolonged corticosteroid therapy, leukemia, end-stage renal disease, chronic malabsorption syndromes) • Children age <4, or those exposed to adults in high-risk categories
≥15 mm	• All of the above plus healthy individuals

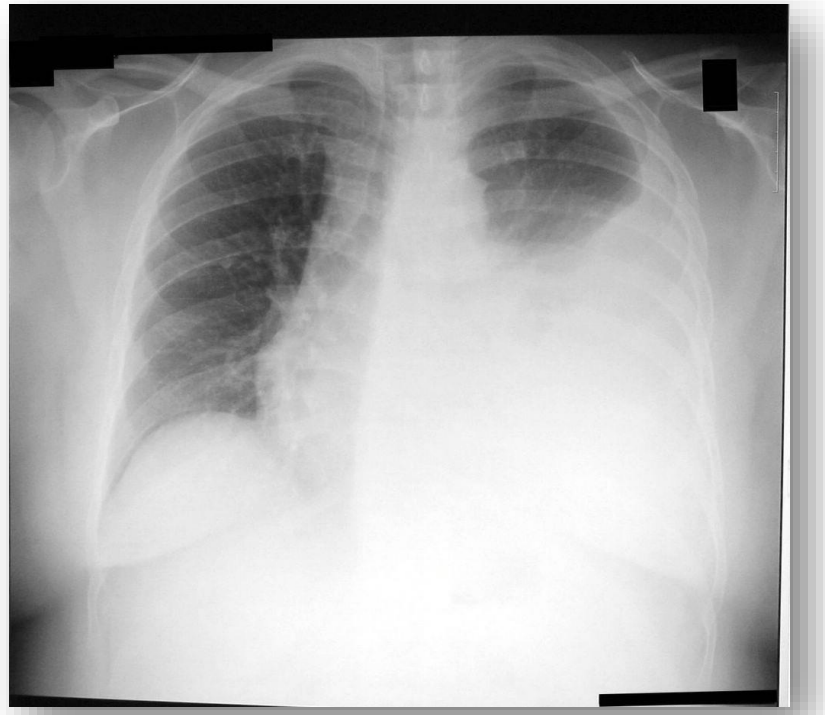
Tuberculosis

- **What's the name of this test ?**
  - Tuberculin skin test
- **What's your diagnosis ?**
  - TB
- **When to interpret the result of this test ?**
  - After 48-72 hours
- **How to measure ?**
  - Depend on the induration not erythema



Tuberculosis

- **Describe what you see in the X-ray ?**
  - Left side homogenous opacity (pleural effusion), with meniscal sign and obliteration of the left costo-phrenic angle and shifting of the mediastinum and heart to the right side.
- **If thoracentesis was done and showed straw colored fluid, what is the causative organism?**
  - Mycobacterium tuberculosis



Respiratory tract infection

Low-Yield

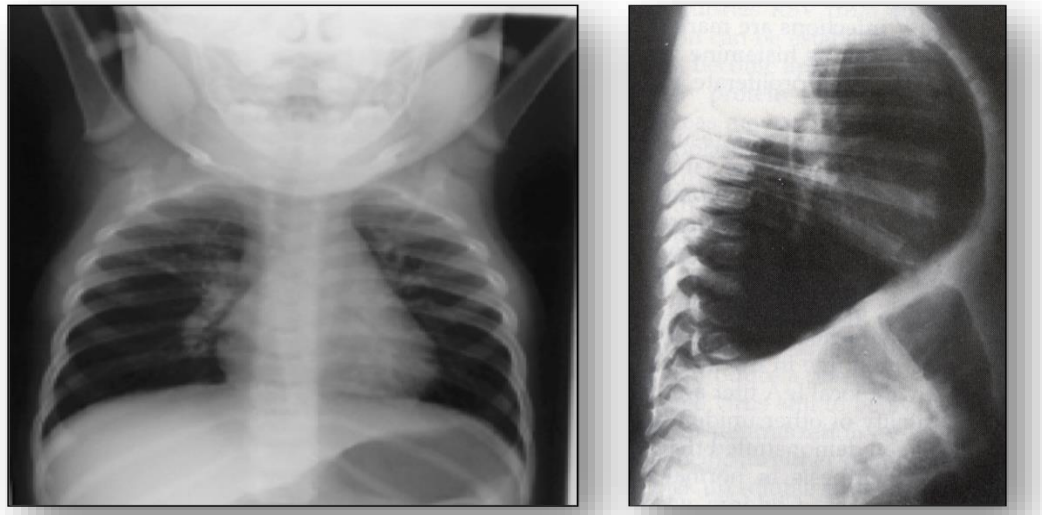
Bronchiolitis

- **Epidemiology**: Primarily affects children < 2 years of age
- **Etiology**: RSV (most common cause), Parainfluenza virus
- **Clinical features**: Initially upper RTI symptoms followed by Lower RTI symptoms (Crackles, wheezes, respiratory distress) and often associated with poor feeding
- **Diagnosis**: Clinically + classic clinical features of bronchiolitis
- **Chest x-ray**: Normal or nonspecific findings e.g., peribronchial thickening, hyperinflation of the lungs, interstitial infiltrates, atelectasis
- **Management**: Supportive measures including O_2 therapy, adequate hydration, relief of nasal congestion and/or obstruction, and monitoring.
- **Complications**: apnea (red flag), super imposed pneumonia, respiratory failure

Bronchiolitis

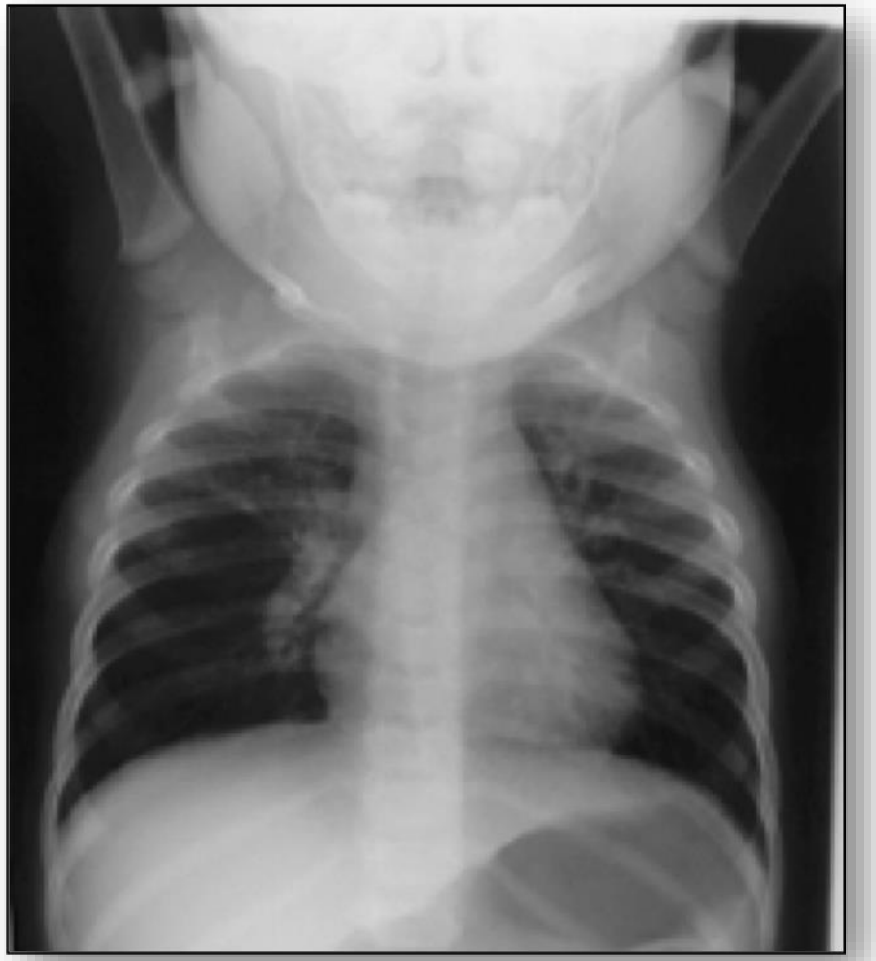
- 3-month-old boy presented to the ER with wheezing and cough and the following x-ray.

- **What do you see in this x-ray and what is your diagnosis?**
  - Hyperinflated chest … Bronchiolitis
- **What is your management ?**
  - Oxygen and fluids



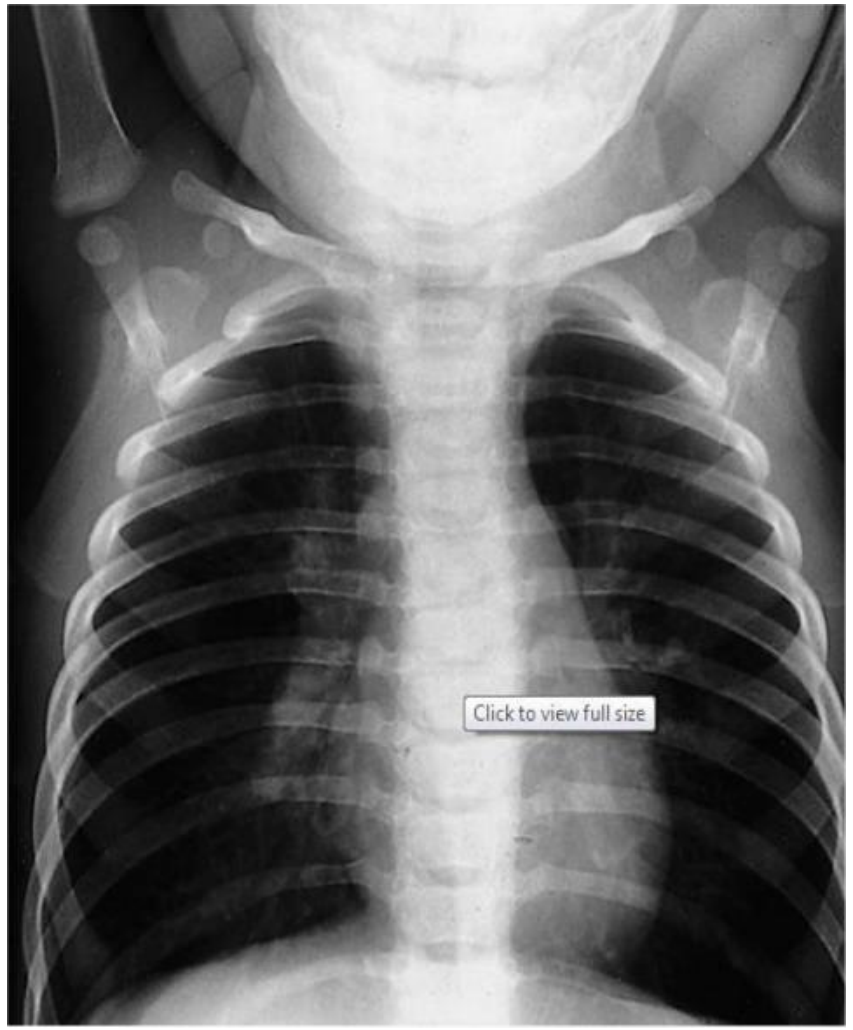
Bronchiolitis

- 7-month-old child, low grade fever, dry cough
- **What are the finding ?**
  - Diffuse pattern of increased interstitial markings, hyperinflation
- **What is causative organism ?**
  - RSV
- **What lab test you'd order ?**
  - Blood gas



Bronchiolitis

- Wheezy chest low grade fever and x ray
- **Mention 2 indications for admission?**
  - hypoxia
  - inability to take oral feedings
  - apnea
  - extreme tachypnea



Croup (Laryngotracheitis, Laryngotracheobronchitis)

- **Epidemiology**: Peak incidence: 6 months to 3 years
- **Etiology**: Parainfluenza viruses (most common cause); RSV
- **Clinical features**: Characteristic features include seal-like barking cough, hoarseness, and inspiratory stridor due to subglottic narrowing
- **Diagnosis**: based on the presence of characteristic clinical features of croup
- **Chest x-ray**: Steeple sign (subglottic narrowing on anteroposterior view)
- **Management**: Start supportive care + Corticosteroid (single dose of Dexamethasone) + Nebulized epinephrine

Croup (Laryngotracheitis, Laryngotracheobronchitis)

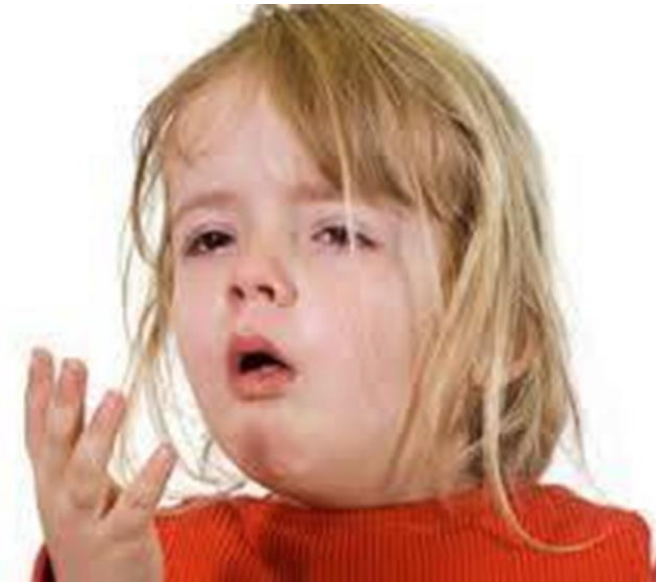
- A patient presents with barking cough and fever he was nontoxic

❖ **What is the diagnosis ?**

- Croup

❖ **What is the most common causative organism ?**

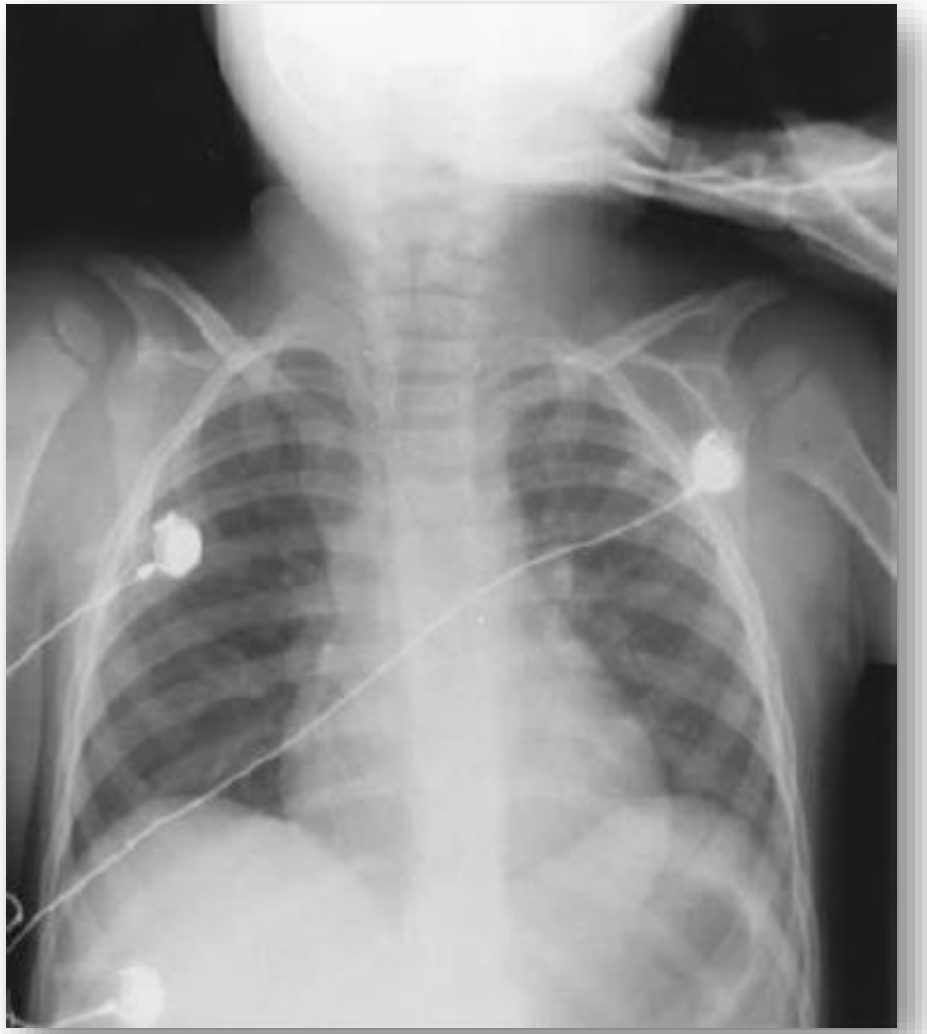
- Parainfluenza virus



Croup (Laryngotracheitis, Laryngotracheobronchitis)

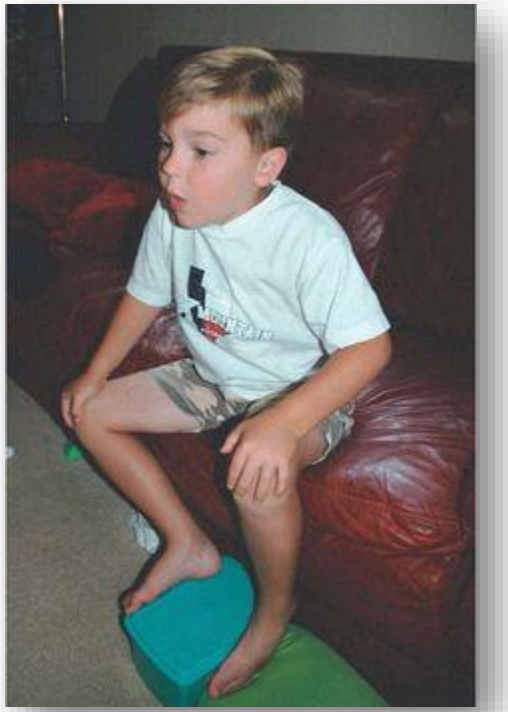
- 1-year-old male presented to the ER with barking cough and respiratory stress with stridor at the rest

- **What is the sign in the X-ray ?**
  - Steeple sign
- **What is the most common cause organism of this condition?**
  - Parainfluenza
- **What is your first line management ?**
  - Start supportive care + Corticosteroid (single dose of Dexamethasone) + Nebulized epinephrine



Epiglottitis

- **Epidemiology**: Peak incidence: 6–12 years
- **What is your diagnosis ?**
  - Epiglottitis
- **What is the most causative organism ?**
  - Haemophilus influenzae type B (Hib)
- **What sign on X-ray does it present with ?**
  - Thumb sign
- **What is the treatment ?**
  - 3rd generation cephalosporin or amoxicillin/clavulanate
- **The hallmarks of epiglottitis are the three Ds:**
  - Dysphagia, Drooling, and Distress.



Tripod position

Pertussis (Whooping cough)

- **Epidemiology**: Typically, a childhood disease (particularly children < 1 year)
- **Etiology**: Bordetella pertussis
- **Stages**:
  - **Catarrhal stage (1–2 weeks)**: Nonspecific symptoms similar to an URTI
  - **Paroxysmal stage (2–6 weeks)**: Intense paroxysmal coughing (often at night)
  - **Convalescent stage (weeks to months)**: Progressive reduction of symptoms
- **Diagnosis**: clinical history and findings
- **Laboratory tests**: lymphocyte-predominant leukocytosis
- **Treatment**: Macrolides (e.g., azithromycin, clarithromycin, erythromycin)

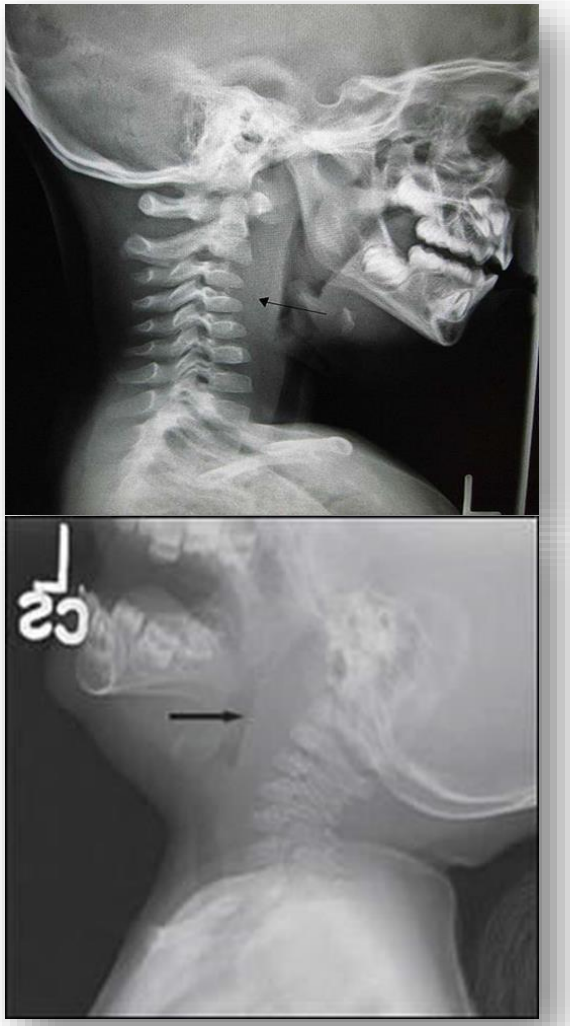
Pertussis (Whooping cough)

- This child did not take any vaccine till this age, now he is presented with paroxysmal cough and coryza

- **What is your spot Dx ?**
  - Pertussis, Whooping cough.
- **What is the cause of this condition ?**
  - Bordetella pertussis.

Child with acute stridor

- **What's the abnormality ?**
  - Increased or swelling of the retro-pharyngeal space
- **What's your Dx.?**
  - Retropharyngeal Abscess
- **What's the treatment?**
  - IV antibiotics & drainage
- **What's the most serious complication for this condition ?**
  - Mediastinitis



Erythema Nodosum

- Hx of boy who had URTI ,then developed bilateral nodular lesion in his legs

❖ **What's your Dx.?**

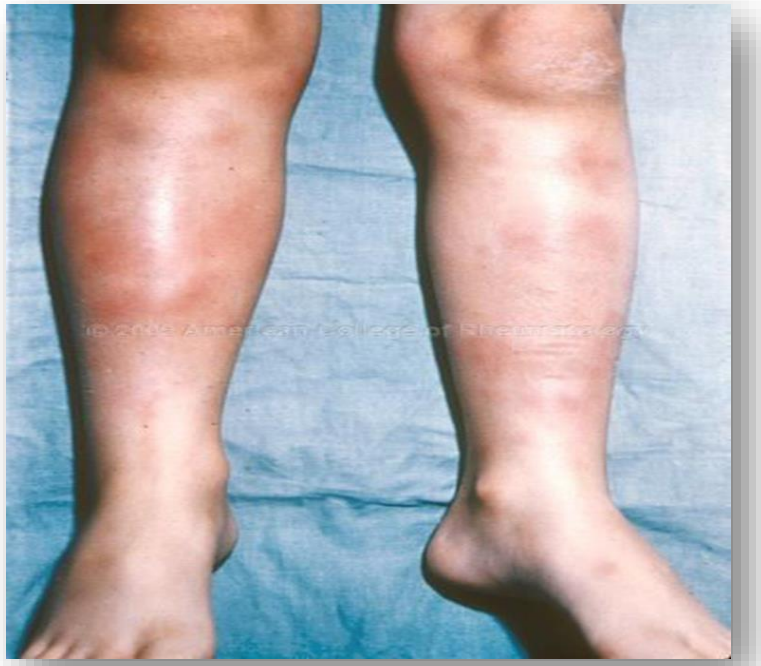
- Erythema Nodosum.

❖ **Give a non-infectious cause.**

- Sarcoidosis.

❖ **Give 2 microorganism causes this condition**

- Group A strep
- Mycoplasma tuberculosis
- Chlamydia



Asthma

Low-Yield

Classification of Asthma

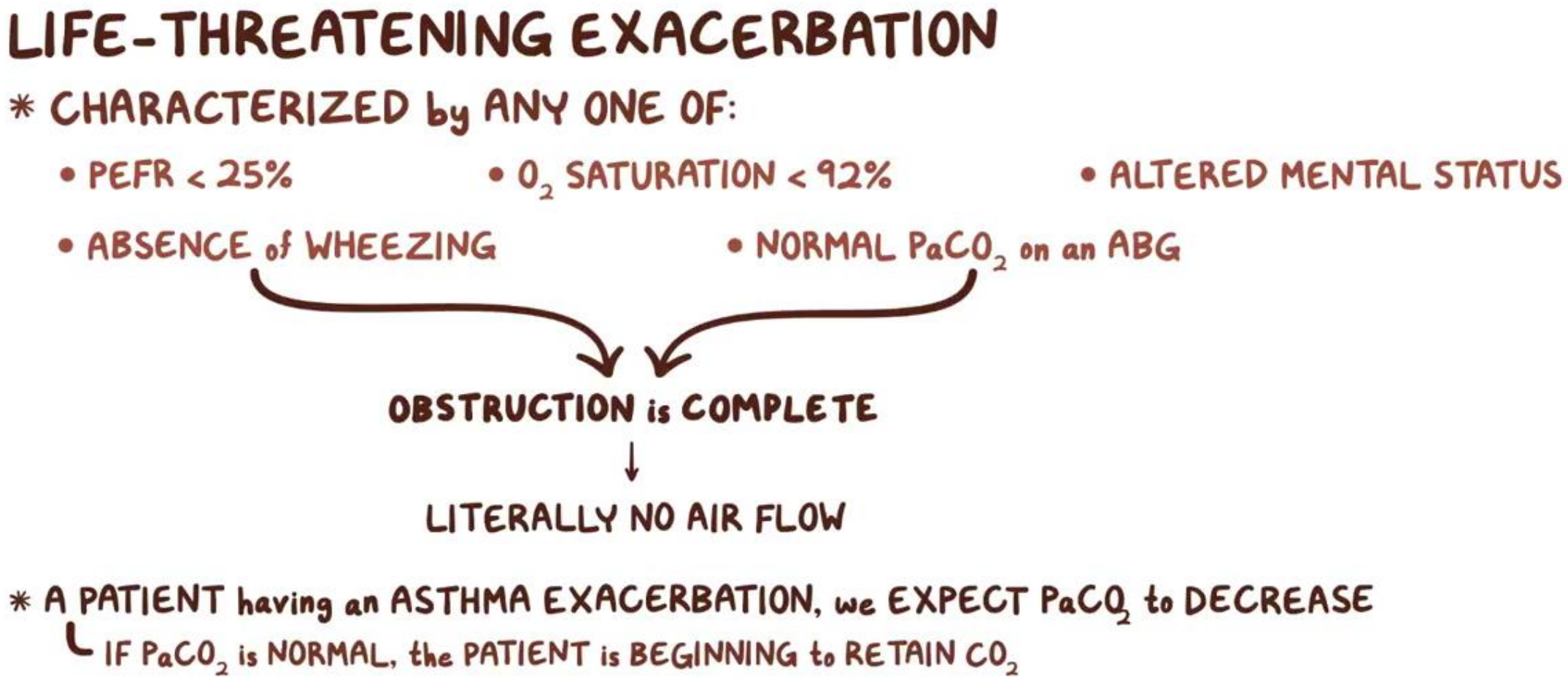
	FREQUENCY DAYTIME SYMPTOMS	FREQUENCY NIGHTTIME SYMPTOMS	FEV1 in between EXACERBATIONS	DEGREE ASTHMA INTERFERES	TREATMENT
INTERMITTENT	< 3 / WEEK	< 3 / MONTH	> 80%	NO LIMITATION	• SABA as needed
MILD PERSISTENT	≥ 3 / WEEK not every day	3 - 4 / MONTH	> 80%	MINOR LIMITATION	• SABA as needed • LOW DOSE INHALED CORTICOSTEROIDS DAILY
MODERATE PERSISTENT	7 / WEEK	1+ / WEEK not every night	60 - 80%	MODERATE LIMITATION	• SABA as needed • LOW DOSE INHALED CORTICOSTEROIDS DAILY • LABA
SEVERE PERSISTENT	THROUGHOUT the DAY	EVERY NIGHT	< 60%	EXTREME LIMITATION	• SABA as needed • LABA • DOSE of INHALED CORTICOSTEROIDS ↑

Asthma exacerbation

	SYMPTOMS	PEFR	ABG
MILD	• ONLY WITH ACTIVITY • GO AWAY AFTER USING an INHALED SABA	> 70%	• USUALLY NOT REQUIRED
MODERATE	• DON'T READILY IMPROVE with SABA	40 - 70%	• USUALLY NOT REQUIRED • LOW $PaCO_2$
SEVERE	ANY ONE of the FOLLOWING • RESPIRATORY RATE > 25 BREATHS/MIN • HEART RATE > 110 BEATS/MIN • PEFR: 25-40% • PATIENT is UNABLE to COMPLETE SENTENCES		• LOW $PaCO_2$ (PATIENT is BREATHING REALLY FAST & BLOWING A LOT of CO_2 OUT)

PARTIAL ARTERIAL PRESSURE of CARBON DIOXIDE ($PaCO_2$)
└ NORMAL: 33 - 45 mm Hg

Asthma exacerbation cont.



Asthma exacerbation cont. 2

NEAR-FATAL EXACERBATION

* MEDICATIONS DON'T WORK
* ↑ $PaCO_2$
* CAN'T BREATHE ON THEIR OWN

↓

VENTILATORY SUPPORT

* CHEST X-RAY
  - USUALLY EITHER NORMAL or SHOWS HYPERINFLATED LUNGS
  - RULE OUT UNDERLYING CAUSE of the ASTHMA EXACERBATION like PNEUMONIA

* DON'T WAIT for ABGs & CHEST X-RAYS *

Asthma exacerbation cont. 3

AFTER PROVIDING TREATMENT in the EMERGENCY DEPARTMENT

PEFR	SYMPTOMS	NEXT STEPS
> 70%	COMPLETELY REVERSED	• PATIENT CAN GO HOME • ARRANGE A FOLLOW-UP PLAN • CHECK if THEY KNOW HOW to USE INHALER
50 -70%	INCOMPLETELY REVERSED	• PATIENT ADMITTED to FLOOR • OBSERVATION • TREATMENT can be CONTINUED
< 50%	NOT IMPROVING	• PATIENT taken to ICU • POSITIVE PRESSURE VENTILATION • INTUBATION

* WHEN DISCHARGING FOLLOWING an EXACERBATION, ALL PATIENTS are PUT ON ORAL CORTICOSTEROIDS for 5 - 10 DAYS

Inhaler spacer

- **What is the name of this device ?**
  - Inhaler spacer
- **What drug is commonly used in it ?**
  - SABA

What are these devices

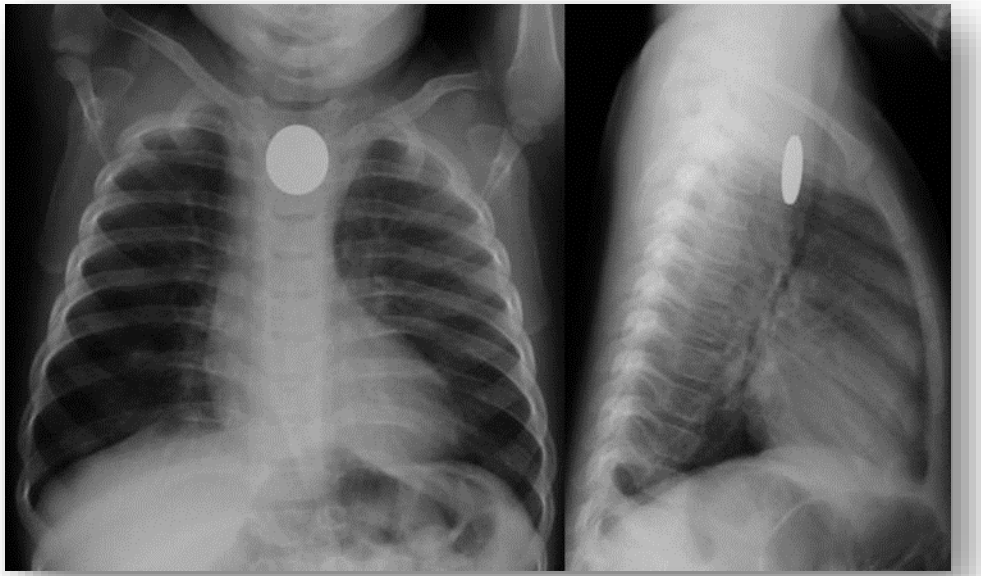
Inhaler

Spacer


ENT
Low-Yield

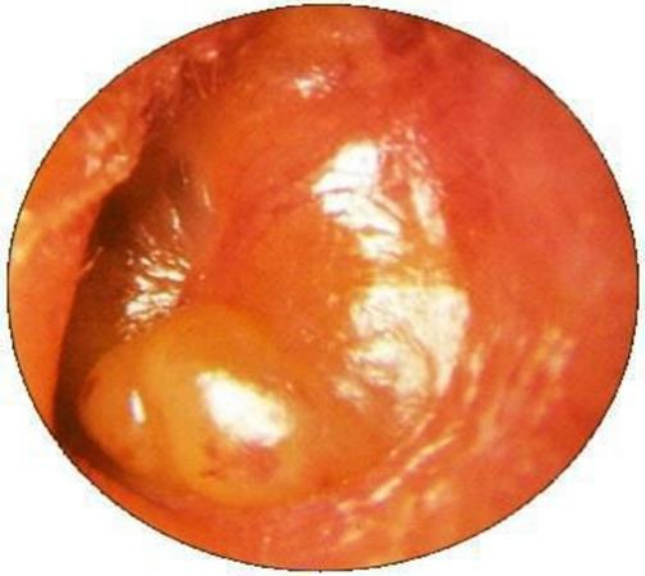
Foreign body ingestion

- 4-year-old Child came with drooling and dysphagia.
- **What is your Dx ?**
  - Foreign body ingestion “coin”
- **What is the management ?**
  - Emergent upper endoscopy



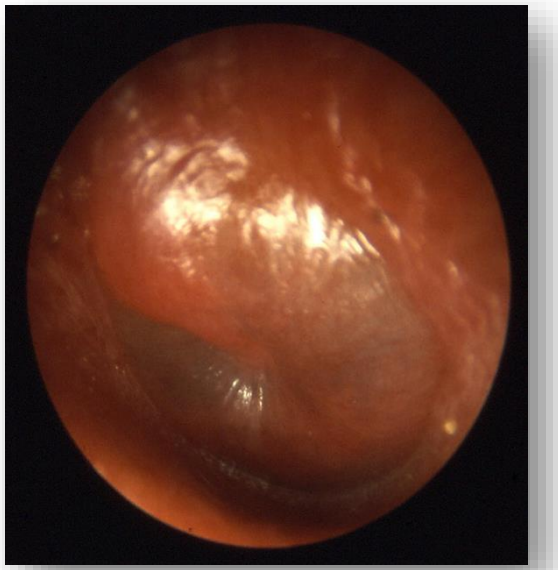
Otitis media with effusion

- **What are 3 findings during ear exam of this patient ?**
  - Erythematous
  - Bulging with fluid
  - Landmarks obscured
  - Likely NOT mobile on pneumatic otoscopy
- **What are the complication of AOM ?**
  - TM perforation
  - Hearing loss
  - Mastoiditis
  - Meningitis
  - Epidural abscess



Otitis media with effusion

- **What's your diagnosis**
  - Otitis media with effusion
- **What's the most common causative organism ?**
  - S.pneumonae
- **Mention 2 risk factors**
  - Adenoid hypertrophy
- **What is the treatment ?**
  - Start Amoxicillin high dose for 7 days



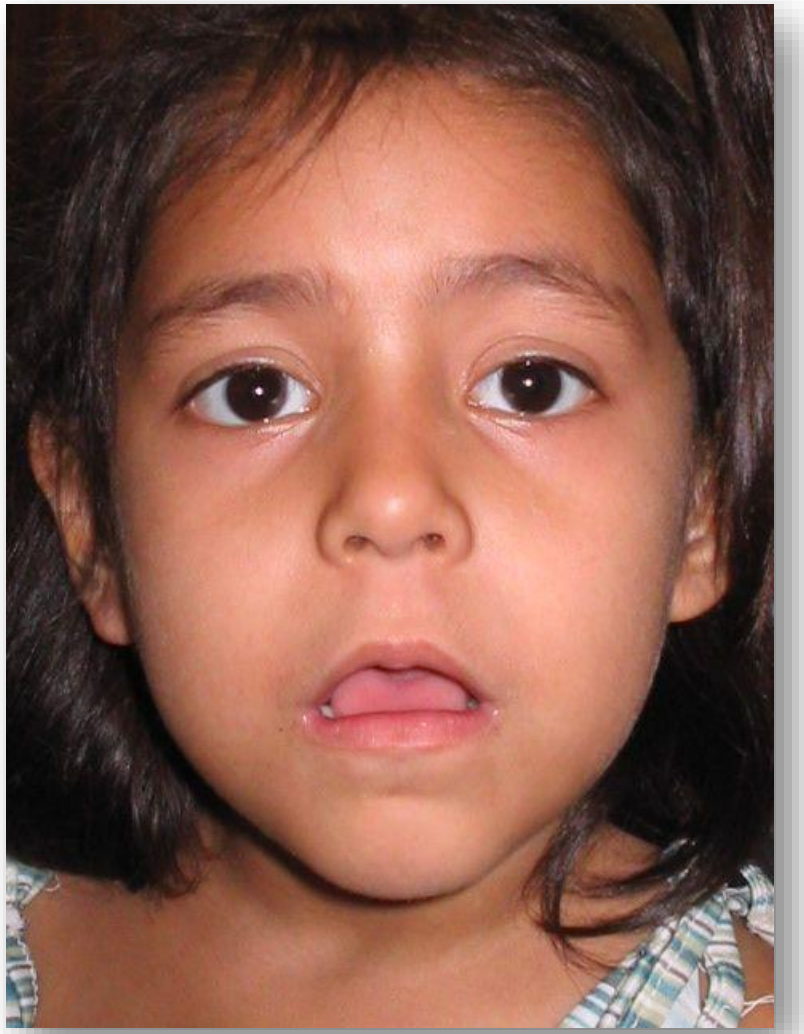
Adenoid hypertrophy

- This kid presented with 2 weeks of rhinorrhea and snoring at night

- **Write 2 physical findings**
  - Mouth breathing + elongated face + the patient looks tired

- **What is your diagnosis ?**
  - Adenoid hypertrophy

- **Write 2 complications of this situation**
  - Middle ear effusion
  - Obstructive sleep apnea
  - Recurrent otitis media with effusion



Pharyngeal tonsils hypertrophy (Adenoids)

❖Symptoms

1. Snoring
2. Difficult noisy breathing
3. Nasal obstruction
4. Nasal discharge
5. Voice change
6. Otitis media with effusion
7. Obstructive sleep apnea

❖What Investigation you should ask for ?

- Post-nasal space X-ray

❖Treatment of adenoid hypertrophy?

- Medical:
  - Anti-histamines
  - Topical nasal steroids.
- Surgical: Adenoidectomy.

Nephrology & Fluids

Nephritic & Nephrotic

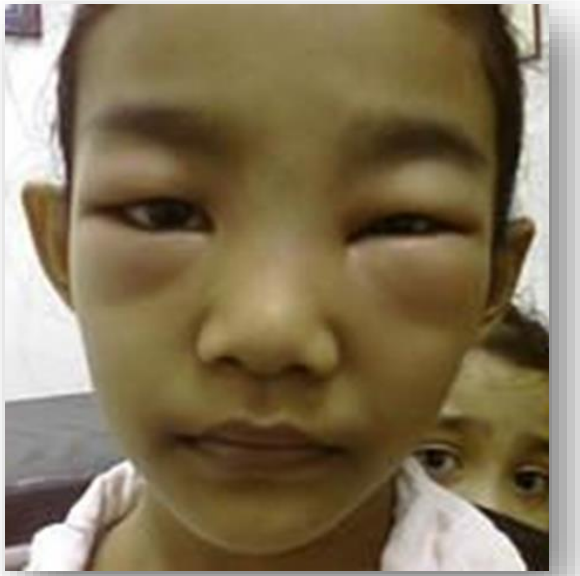
وحاجات حلوة أخرى

Medium-Yield

إضافي

TYPE	ETIOLOGY	CLINICAL PRESENTATION	EXAMPLES
Nephritic syndrome	Glomerular inflammation → GBM damage → loss of RBCs into urine → dysmorphic RBCs, hematuria	Hematuria, RBC casts in urine ↓ GFR → oliguria, azotemia ↑ renin release, HTN Proteinuria often in the subnephrotic range (< 3.5 g/day) but in severe cases may be in nephrotic range	▪ Infection-associated glomerulonephritis ▪ Goodpasture syndrome ▪ IgA nephropathy (Berger disease) ▪ Alport syndrome ▪ Membranoproliferative glomerulonephritis
Nephrotic syndrome	Podocyte damage → impaired charge barrier → proteinuria	Massive proteinuria (> 3.5 g/day) with edema, hypoalbuminemia → ↑ hepatic lipogenesis → hypercholesterolemia Frothy urine with fatty casts Associated with hypercoagulable state due to antithrombin III loss in urine and ↑ risk of infection (loss of IgGs in urine and soft tissue compromise by edema)	May be 1° (eg, direct podocyte damage) or 2° (podocyte damage from systemic process): ▪ Focal segmental glomerulosclerosis (1° or 2°) ▪ Minimal change disease (1° or 2°) ▪ Membranous nephropathy (1° or 2°) ▪ Amyloidosis (2°) ▪ Diabetic glomerulonephropathy (2°)
Nephritic-nephrotic syndrome	Severe GBM damage → loss of RBCs into urine + impaired charge barrier → hematuria + proteinuria	Nephrotic-range proteinuria (> 3.5 g/day) and concomitant features of nephritic syndrome	Can occur with any form of nephritic syndrome, but is most common with: ▪ Diffuse proliferative glomerulonephritis ▪ Membranoproliferative glomerulonephritis

Hx of URTI 10 days ago, BP was normal



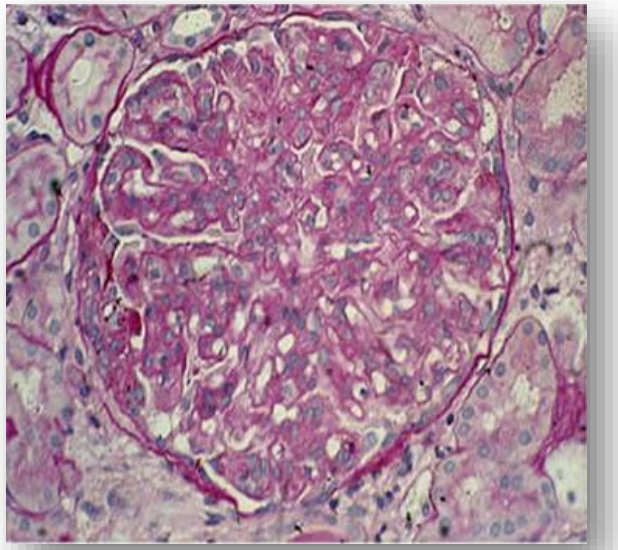
- **Describe what you see.**
  - Bilateral peri-orbital edema.
- **What 1st & rapid test you want to do?**
  - Urine dipstick
- **Mention 2 other things you want to examine.**
  - Lower limb edema
  - Chest (pleural effusion)
  - Abdomen “ascites”

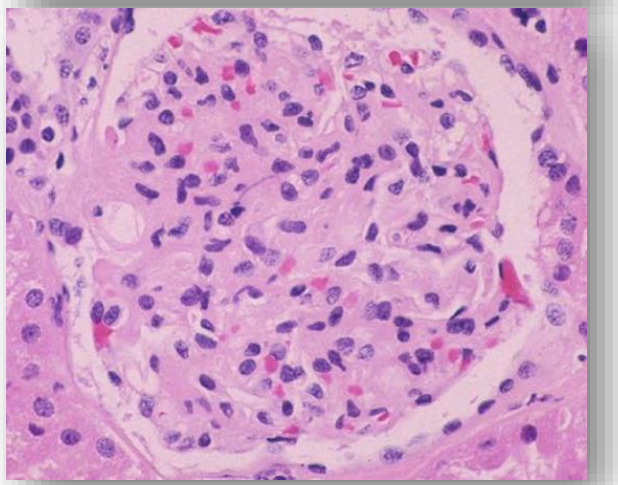
➢ 2 weeks ago, this boy started to develop this clinical picture.

- **Mention 2 tests you will order to support your diagnosis.**
  - Urinalysis
  - Antistreptococcal antibody titers (ASO, Anti-DNase B), C3 complement level

Hematuria and Hx of URTI 2 weeks ago

- **Mention 2 ddx?**
  - PSGN
  - IgA nephropathy
- **Mention 2 investigations to confirm your Dx**
  - C3 complement level, anti-DNAase, ASO
- **Mention 2 acute complications**
  - HTN, fluid overload
- **What other symptoms or sign you can see in this patient ?**
  - Oliguria, Azotemia, HTN, Mild to moderate edema





Red urine 2 days after URTI

- **What is your diagnosis ?**
  - IgA nephropathy
- **Mention 2 abnormalities in lab tests associated with this condition**
  - Serum IgA level is elevated in 50% of patients
  - Complement levels (e.g., C3 level) are generally normal

Nephrotic syndrome

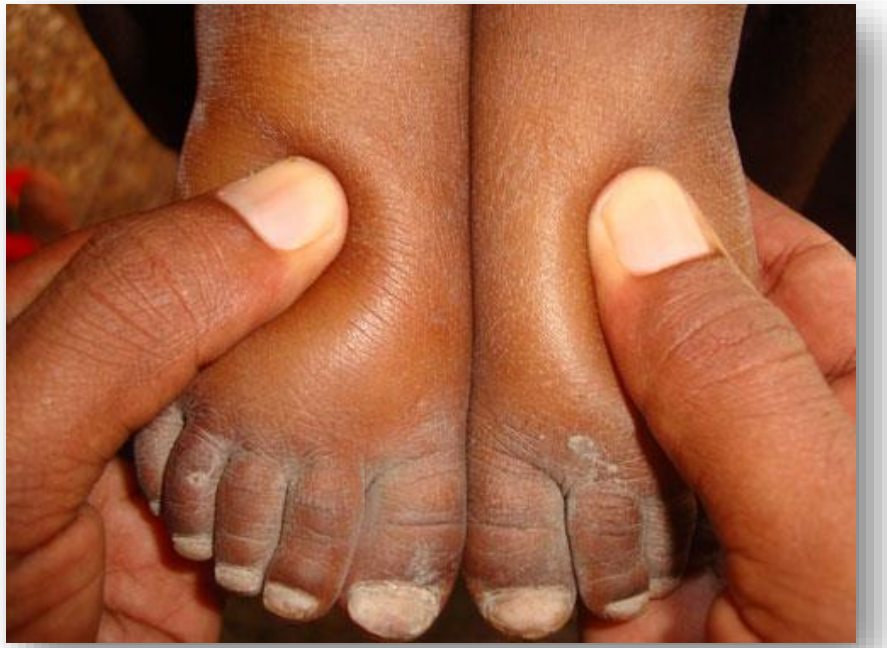
- This child come with 2-day history of facial puffiness
- **What is your differential diagnosis ?**
  - Minimal change disease, trauma, insect bite
- **What the investigations that you can use to confirm your diagnosis ?**
  - 24 urine protein collection
  - Lipid profile
  - Albumin level

Patient known to have nephrotic syndrome

- **Mention 3 lab tests to confirm the diagnosis**
  - 24 urine protein collection
  - Lipid profile
  - Albumin level

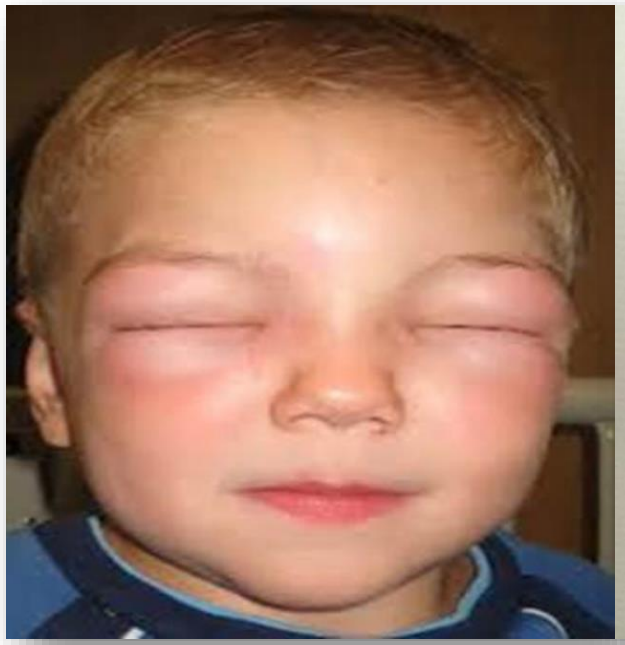
Periorbital and limb edema

- **Mention 2 possible complications**
  - HTN, fluid overload, hyperkalemia, hyponatremia, acidosis, thrombosis, Spontaneous bacterial peritonitis
- **Mention 3 lab tests to confirm the diagnosis**
  - 24 urine protein collection
  - Lipid profile
  - Albumin level



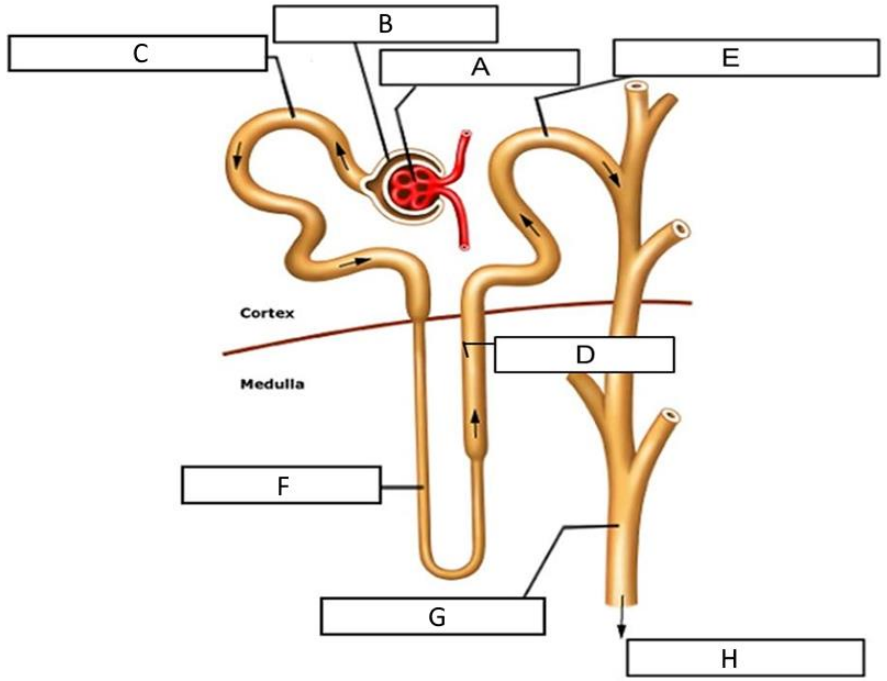
Nephrotic-nephritic presentation

- **What's your diagnosis ?**
  - Nephrotic-nephritic presentation “hematuria”
- **Mention two lab tests to support your diagnosis ?**
  - Urine analysis, Serum albumin level, 24-hour urine collection



Nephron

- **What is the name of the structure labeled as A ?**
  - Glomerulus
- **What Is the site of action of Furosemide ?**
  - D (Loop of henle)
- **What diuretic acts on part E ?**
  - Thiazides



Preauricular Sinus

- After doing hearing tests, what's the next step you must do?
  - Renal US, KFT.

UTI, VUR & Enuresis

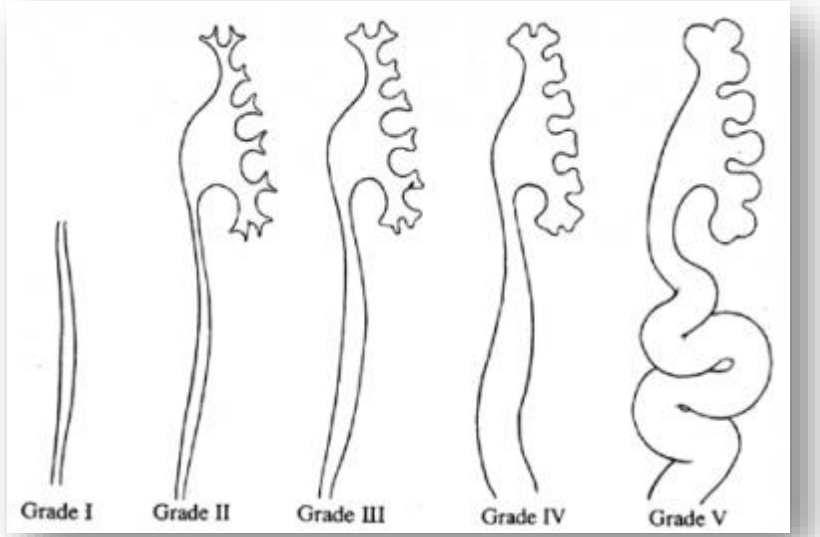
Medium-Yield

Urine dipstick

- **Mention 2 findings in dipstick are useful to diagnose UTI**
  - Leukocyte esterase
  - Alkaline pH
  - Nitrites
  - RBC

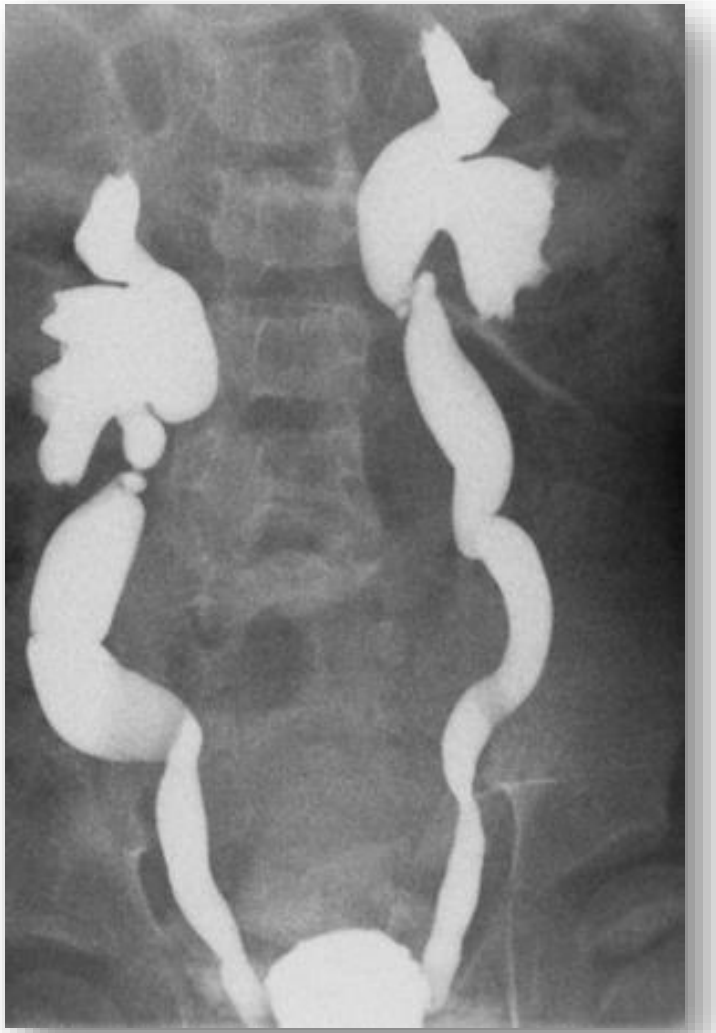
Vesicoureteral Reflux Grading

- **Grade I**: Urine flow back into on or both of the ureters but doesn't reach the kidney (urine reflux into non-dilated ureters)
- **Grade II**: Flow back up to the kidney but doesn't cause dilatation of renal pelvis
- **Grade III**: Mild to moderate dilatation of the ureter, renal pelvis and calyces with minimal blunting at fornices
- **Grade IV**: Moderate to severe dilatation of the ureter, renal pelvis and calyces with mild tortuosity
- **Grade V**: Severe dilatation with severe tortuosity, blunting renal fornices, cortical thinning and ballooning



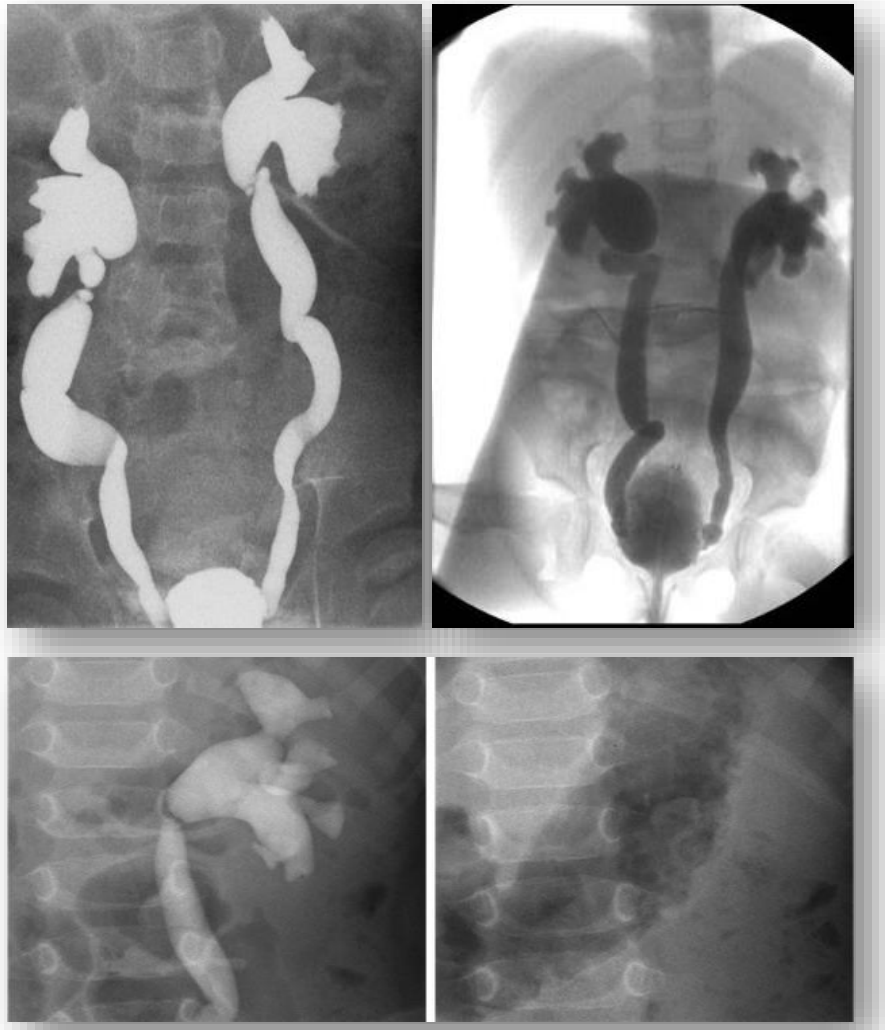
Vesicoureteral reflux

- **What is this technique called ?**
  - MCUG or VCUG (Voiding cystourethrogram)
- **Mention 2 possible causes**
  - Vesicoureteral reflux
  - Posterior urethral valve
  - Neurogenic bladder



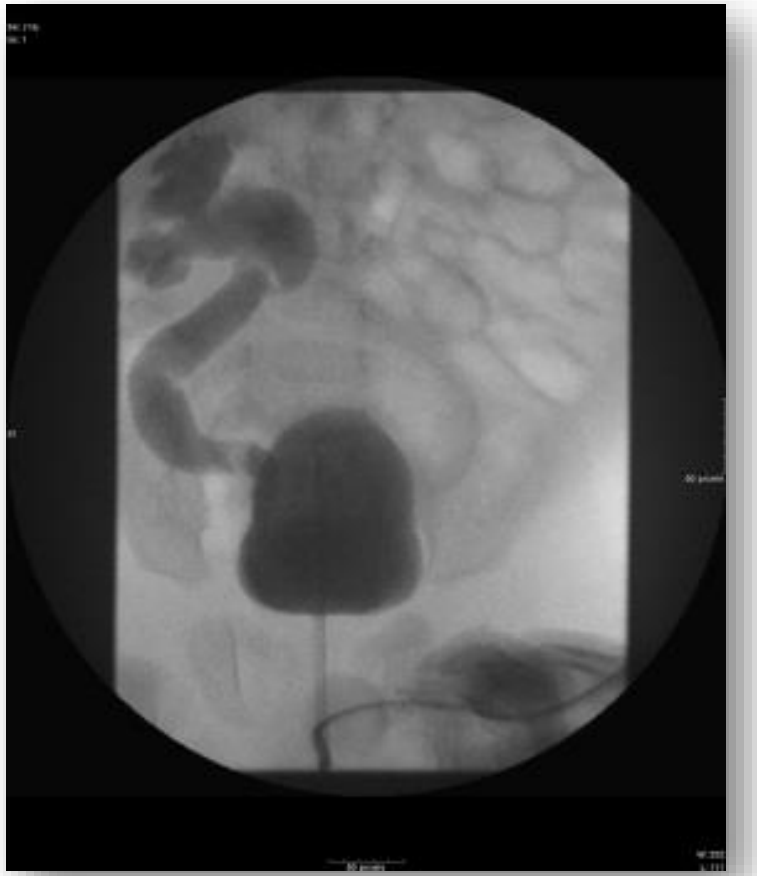
Vesicoureteral reflux

- **What is this technique called ?**
  - MCUG or VCUG (Voiding cystourethrogram)
- **What are the findings is the photo ?**
  - Dilation in ureters
  - Dilation of renal pelvis and calyces
  - Mild tortuosity on the left
- **Write down 2 complications ?**
  - Recurrent UTI
  - Reflux nephropathy



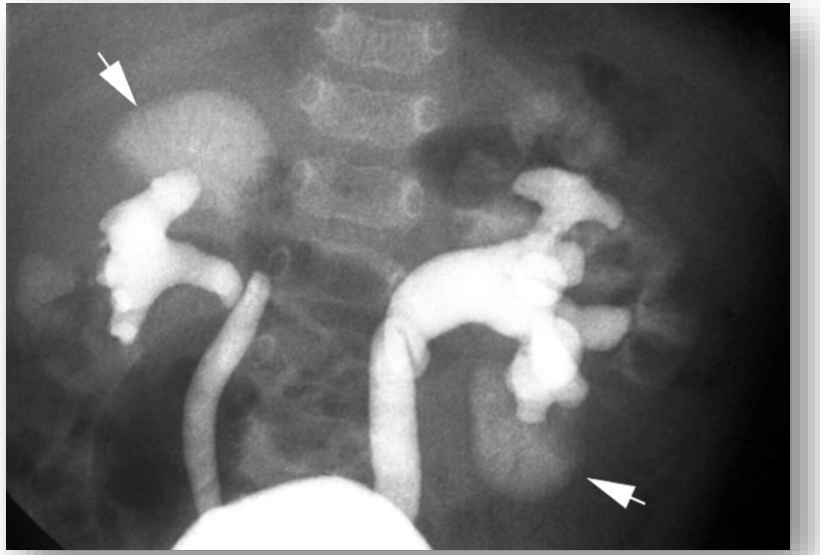
Vesicoureteral reflux

- **What are the findings is the photo ?**
  - Hydro-uretro-nephrosis & ureter touristy
- **Write down 2 complications ?**
  - Recurrent UTI
  - Reflux nephropathy



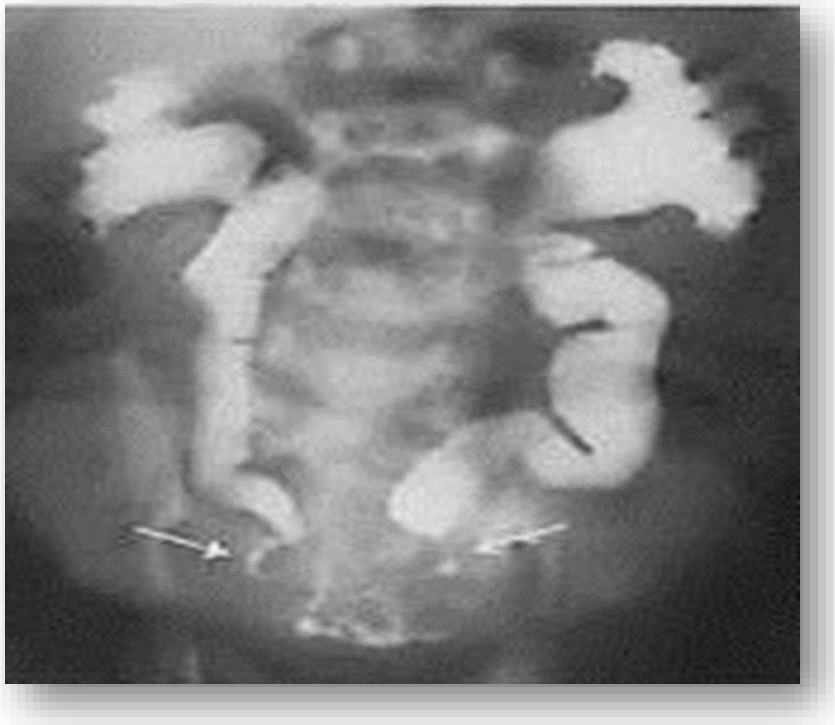
Vesicoureteral reflux

- **What is this technique called ?**
  - MCUG or VCUG (Voiding cystourethrogram)
- **What is your diagnosis?**
  - Vesicoureteral reflux
- **What is the usual presentation of this disease?**
  - Recurrent UTI



Vesicoureteral reflux

- **What's the name of this test?**
  - VCUG or MCUG.
- **What's your next test?**
  - Late DMSA scan.



Vesicoureteral reflux

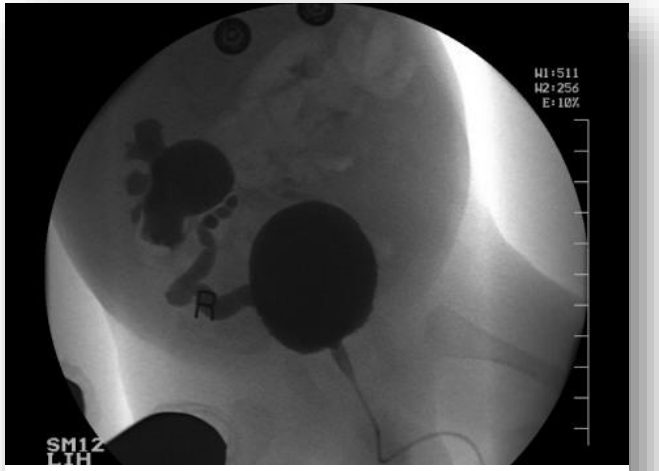
- 12-week-old boy is admitted with a fever and being generally unwell. He is found to have a UTI. He is treated with antibiotics and several weeks after the infection resolves he has several investigations performed

❖ **What kind of investigation is radiological investigation 1 and what does is show ?**

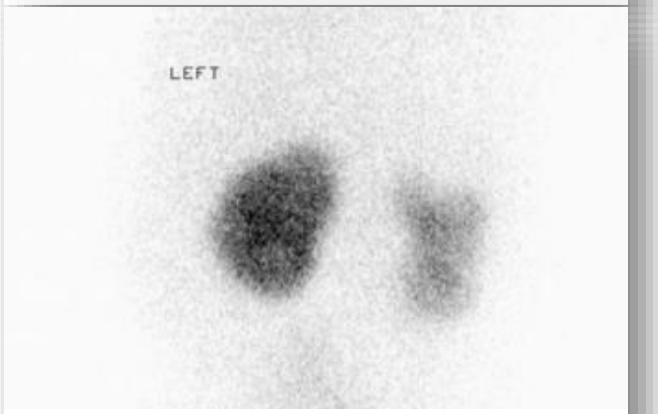
- Micturating cystourethrogram (MCUG). This shows vesicoureteric reflux on the right side with right sided hydroureter and hydronephrosis

❖ **What kind of investigation is radiological investigation 2 and what does is show ?**

- A nuclear medicine investigation called a DMSA renal scan. It shows that the right kidney is small and scarred with lesions at the upper pole and low/mid lateral territory



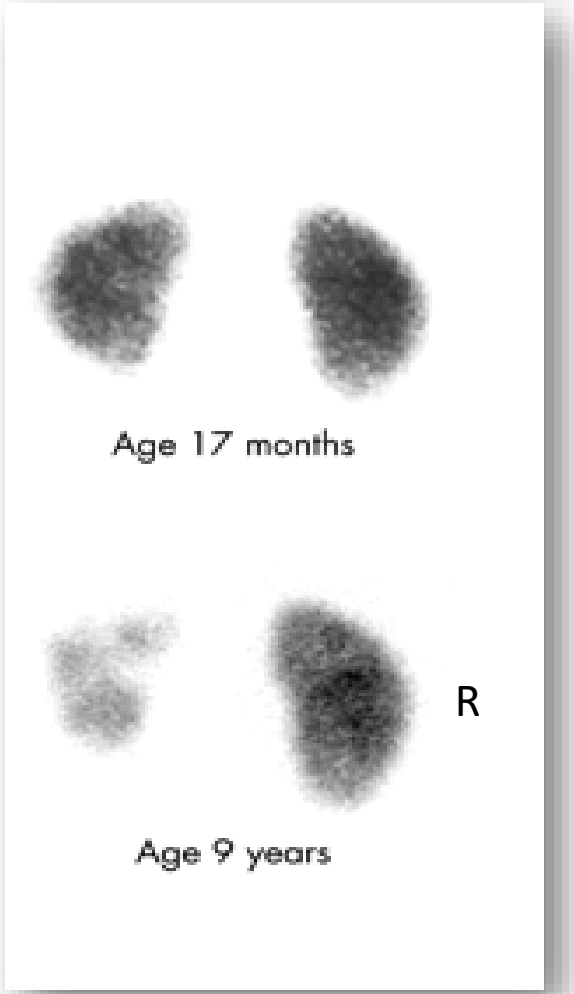
Radiological investigation 1



Radiological investigation 2

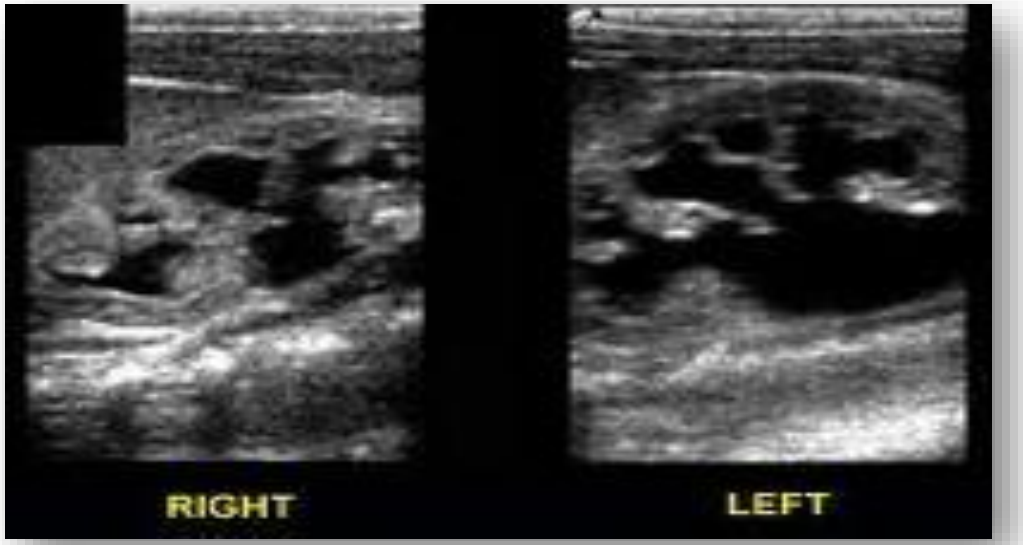
Kidney Scaring

- **What are the findings in this DMSA scan image ?**
  - Severe left kidney scarring which is progressive over the period between 17 months and 9 years
- **Mention 2 possible complications**
  - ESRD, acidosis, volume overload, elec. disturbance .. etc (not sure)
- **What is the cause ?**
  - Reflux nephropathy
- **Other tests to do ?**
  - Biopsy, KFT



Hydronephrosis

- **Mention 2 causes**
  - Vesicoureteral reflux
  - Neurogenic bladder
  - Posterior urethral valve
- **What is the next investigation**
  - VCUG



Enuresis

- **Enuresis**: Urinary incontinence > **2** times / week for > **3** months in persons > **5** years old
- Treatment of enuresis is not indicated before 5 years of age as most cases resolve spontaneously!
- **The first line treatments** for Enuresis are behavioral modification (e.g., Fluid restriction at night) and positive reinforcement
- **For refractory cases** of Enuresis, you can use a bedwetting alarm or oral desmopressin
- Imipramine can be used, but desmopressin is MUCH more preferred due to more favorable side effect profile

Chronic Kidney disease

Low-Yield

إضافي

Consequences of renal failure

Decline in renal filtration can lead to excess retained nitrogenous waste products and electrolyte disturbances.

Consequences (**MAD HUNGER**):

- **M**etabolic **A**cidosis
- **D**yslipidemia (especially ↑ triglycerides)
- **H**igh potassium
- **U**remia
- Na^+/H_2O retention (HF, pulmonary edema, hypertension)
- **G**rowth retardation and developmental delay
- **E**rythropoietin deficiency (anemia)
- **R**enal osteodystrophy

2 forms of renal failure: acute (eg, ATN) and chronic (eg, hypertension, diabetes mellitus, congenital anomalies).

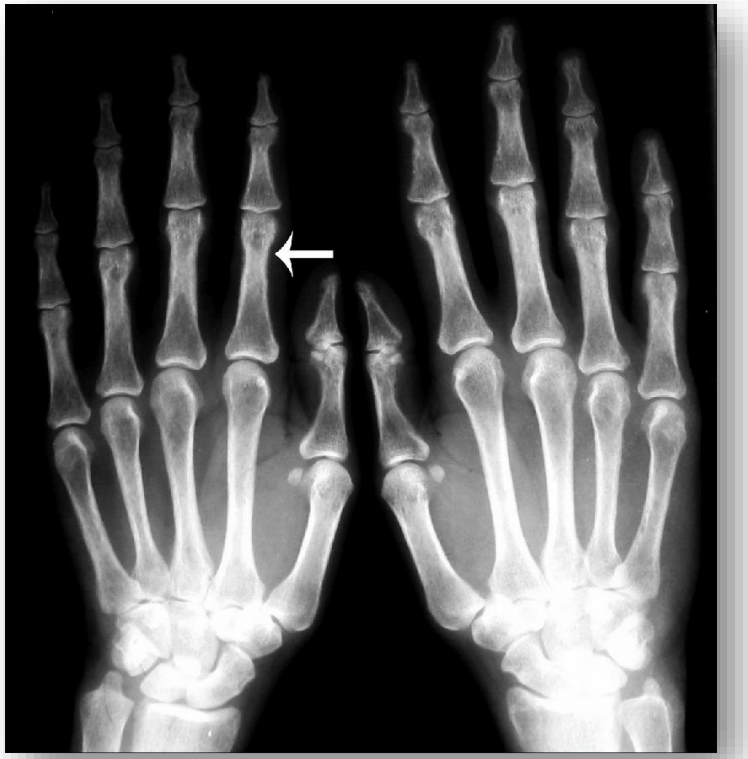
Incremental reductions in GFR define the stages of chronic kidney disease.

Normal phosphate levels are maintained during early stages of CKD due to ↑ levels of fibroblast growth factor 23 (FGF23), which promotes renal excretion of phosphate. "**F**GF23 **f**ights **f**(ph)osphate."

Uremia—syndrome resulting from high serum urea. Can present with **P**ericarditis, **E**ncephalopathy (seen with asterixis), **A**norexia, **N**ausea (pronounce "**Ure-PEAN**" [European]).

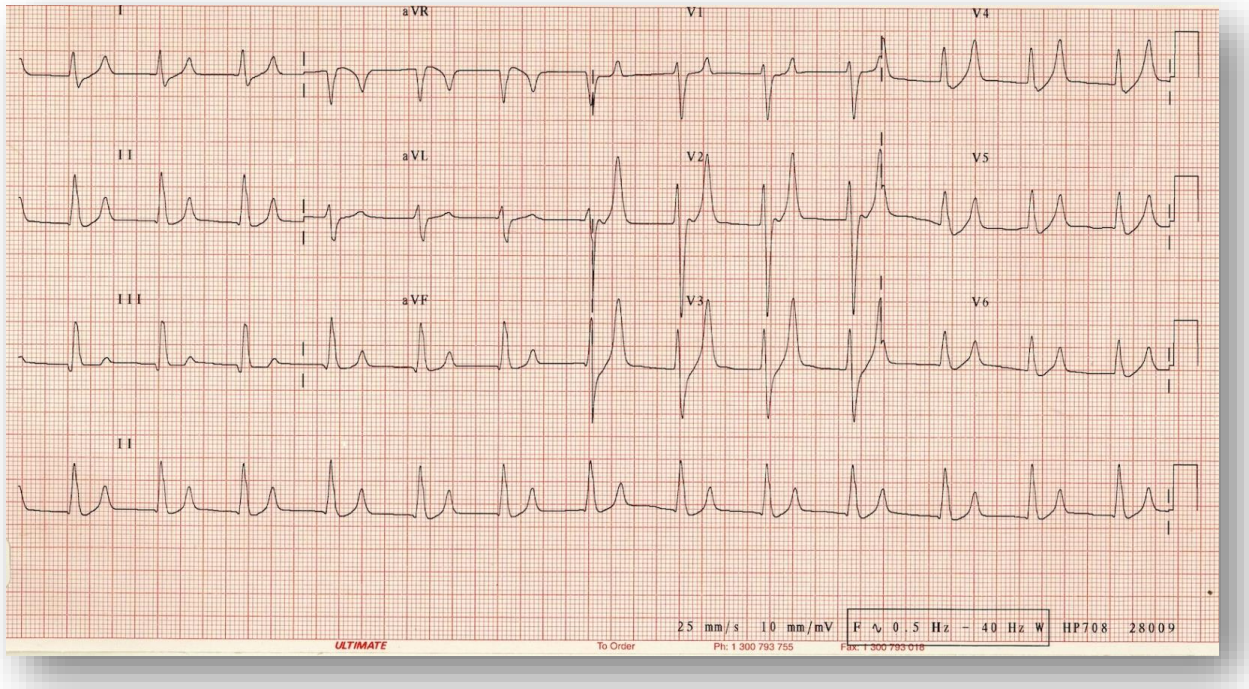
Renal osteodystrophy

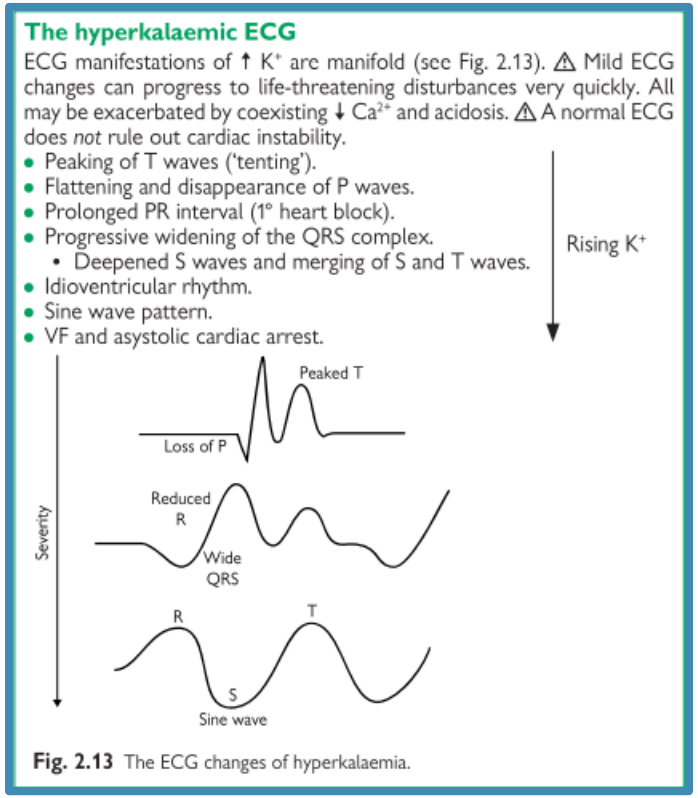
- 15-year-old male presented with bone pain from months: high urea and creatinine, eGFR=40
- **What is the Name of this condition ?**
  - Metabolic bone disease (renal osteodystrophy)
- **Mention 2 lines of treatment**
  - Calcitriol (active form vit. D) + control of phosphate level (restrictive of phosphate intake and give phosphate binders)
- **Write 2 complications of this situation**
  - Anemia, volume overload (cause CHF and death), metabolic acidosis (high anion gap)



Patient with chronic renal failure presented with this ECG

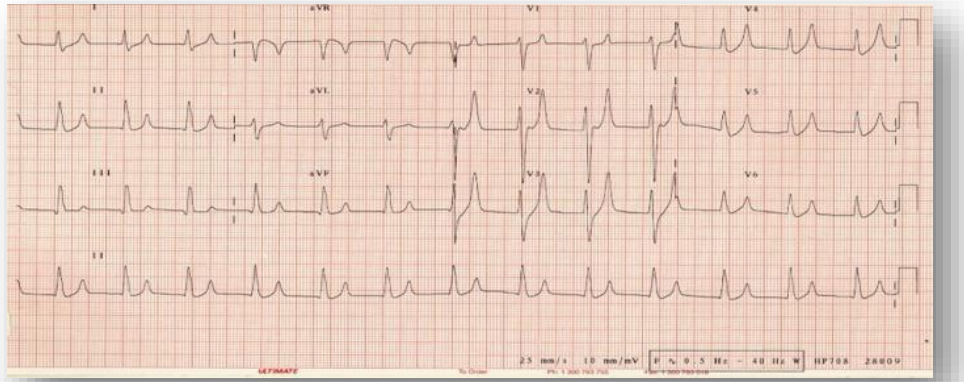
- **What immediate test you should do for this patient ?**
  - Serum potassium level
- **What immediate drug you should give ?**
  - Calcium gluconate (indications: K >6.5 or ECG changes)

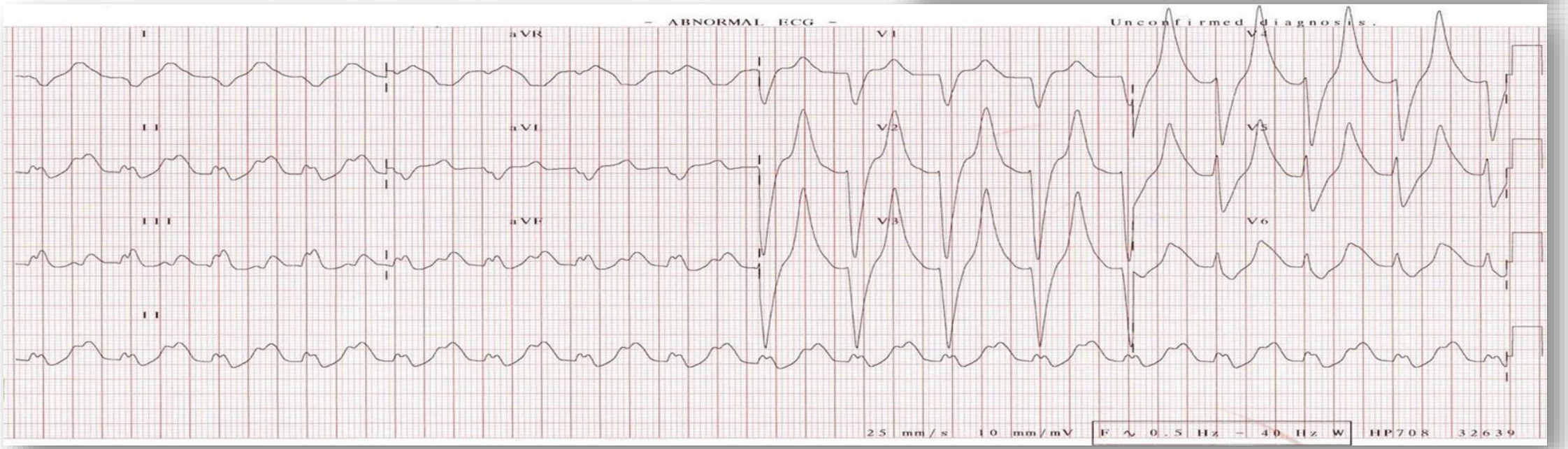




Hyperkalemia

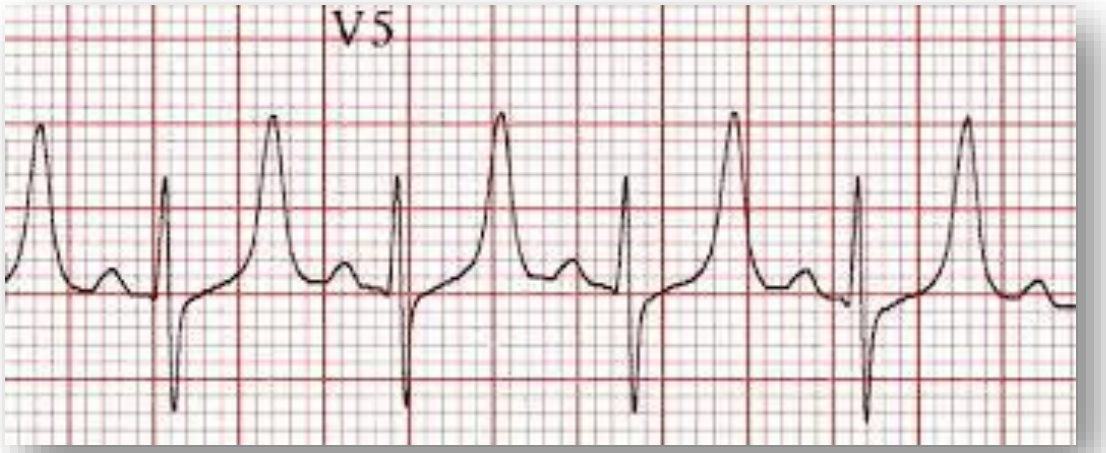
- **Name the finding on ECG**
  - Peaked T wave
- **Give 3 modalities for treatment**
  - Calcium gluconate, Insulin, β2-agonist





Hyperkalemia

- **What is the electrolyte abnormality in this ECG?**
  - Hyperkalemia
- **What is the first line management?**
  - Ca gluconate
- **All of the following are causes of this ECG change except**
  - Pyloric stenosis
  - Addison disease
  - Rhabdomyolysis
  - Labetalol
  - Captopril



Management of hyperkalemia

- **Calcium gluconate**
  - **Specific Indication**: K^+ >6.5 mmol/L or ECG changes
  - **MOA**: Cardioprotective, doesn't decrease K^+ levels
  - Give over 2-5 min repeat after 5-minute if there is no ECG improvement
- **Insulin & Glucose**
  - **Specific Indication**: K^+ >6.5 mmol/L or ECG changes
  - **MOA**: Increase activity of Na/K ATPase moving the K^+ from the plasma to the cells
- **Sodium bicarbonate**
  - **Specific Indication**: High K^+ levels in the presence of acidosis and hypovolemia
  - **MOA**: Increase activity of Na/K ATPase
- **β2-agonist**
  - **MOA**: Increase activity of Na/K ATPase
- Urinary K^+ wasting: by using diuretics; contraindicated in renal problems
- Gut K^+ wasting: by using cation exchange resins

Fluids

جاء بس سؤال واحد مرة وحدة فمش رح رجع أكتب محاضرة مشانه بعينكم الله حبايبي

Low-Yield

Dehydration degree			
Percentage of TBW	**Mild**	**Moderate**	**Severe**
% in infants	5% (50 mL/kg)	10% (100 mL/kg)	15% (150 mL/kg)
% in adolescents	3% (30 mL/kg)	6% (60 mL/kg)	9% (90 mL/kg)
Clinical signs			
Systemic signs	Increase thirst	Irritable	Lethargic
Urine output	Decreased	<1mL/Kg/hr	Oliguria or Anuria
Mucus membrane	Tachy (Sticky)	Dry	Parched
Skin turgor*	Normal	Reduced	Tenting
Capillary refill*	Normal	Mildly delayed	Markedly delayed
Skin temperature	Normal	Cool	Cool, mottled
Anterior fontanel	Normal	Sunken	Markedly sunken
Heart rate	Normal	Increased	Markedly increased or ominously low
Blood pressure	Normal	Normal to low	Low
Respirations*	Normal	Deep, maybe increased	Deep and increased or decreased to absent

Prolonged capillary refill, Abnormal skin turgor, Abnormal respiration *: Best predicators of at least 5% dehydration

Oral rehydration therapy

- Used in mild to moderate dehydration
- **Step 1**: Deficit replacement
  - 50mL/Kg over 4hr. If mild
  - 100mL/Kg over 4hr. If moderate
- **Step 2**: Replace ongoing losses
  - **If vomiting**: 2mL/Kg of ORS after each episode of vomiting
  - **If diarrhea**: 10mL/Kg of ORS after each diarrheal stool
- **Step 3**: Maintenance
  - When rehydration is complete, maintenance therapy should be started, using 100 mL of ORS/kg in 24 hours until the diarrhea stops.
  - Breast feeding or formula feeding should be maintained and not delayed for more than 24 hours.
  - Children should continue regular diet (no bowel rest), but **restrict** juices and sugary drinks (ممنوع العصائر)

Oral rehydration therapy

- **When should you switch to IV fluid therapy ?**
  - Circulatory instability or shock
  - Altered mental status
  - Intractable vomiting
  - Bloody diarrhea
  - Ileus
  - Abnormal serum Na
  - Glucose malabsorption

Concepts of rehydration therapy

1. **Maintenance**: Normal daily need of fluids and electrolytes
   - **Fluid maintenance**: Holliday-Segar Method (100-50-20 rule)
     - **First 10Kg**: 100mL/Kg/day or 4mL/Kg/hour
     - **Second 10Kg**: 50mL/Kg/day or 2mL/Kg/hour
     - **Additional Kg**: 20mL/Kg/day or 1mL/Kg/hour
   - **Solutes Maintenance**
     - **Na^+ maintenance**: 3mEq/100mL of maintenance fluid (i.e., if the child need 1L of fluid he will need 30mEq of Na)
     - **K^+ maintenance**: 2mEq/100mL of maintenance fluid (i.e., if the child need 1L of fluid he will need 20mEq of K)

Concepts of rehydration therapy

2. **Deficit**: Losses of fluids and electrolytes resulting from an illness
   - **Fluid deficit**: % of dehydration * body weight (ideally before illness)
     - E.g., child weight 7Kg with 10% dehydration: 7000g * 0.1 = 700mL
   - **Solutes deficit**: Fluid deficit (L) * Solute mEq/100ml * % according to the following table
     - **Na during first 3 days**: Fluid deficit (L) * 140 * 0.8 = 11.2 * Fluid deficit (L)
     - **Na after first 3 days**: Fluid deficit (L) * 140 * 0.6 = 8.4 * Fluid deficit (L)
     - **K during first 3 days**: Fluid deficit (L) * 150 * 0.2 = 3 * Fluid deficit (L)
     - **K after first 3 days**: Fluid deficit (L) * 150 * 0.4 = 6 * Fluid deficit (L)

ECF deficit (mainly Na)	ICF deficit (mainly K)
<3 days = 80%	<3 days = 20%
≥3 days = 60%	≥3 days = 40%

Concepts of rehydration therapy

- **Note**: Emergency bolus
  - The child is given a fluid bolus, usually 20 mL/kg of the isotonic solution, over about 20 minutes.
  - To ensure that the intravascular volume is restored, the patient receives an additional 20 mL/kg bolus of isotonic fluid over 2 hours.
  - The child's total fluid needs are added together (maintenance + deficit).The remaining fluid volume is then administered over 24 hours.
  - Potassium usually is not included in the IV fluids until the patient voids, unless significant hypokalemia is present.
  - Children with significant ongoing losses need to receive an appropriate replacement solution

Types of dehydration

Types of dehydration	Sodium (mEq/L)	Common causes
Isonatremic	130-150	Secretory diarrhea
Hyponatremic	< 130	Replacing with hypotonic fluids
Hypernatremic	> 150	Viral gastroenteritis, e.g. rotavirus

Calculations

Steps	Isonatremic	Hyponatremic	Hypernatremic
Step 1: Maintenance	Maintenance of fluids and electrolytes for 24hrs.	Maintenance of fluids and electrolytes	Maintenance of fluids and electrolytes for 48hrs.
Step 2: Deficits	Deficits of fluids and electrolytes	Deficits of fluids and electrolytes	Deficits of fluids
		Additional Na deficit • mEq Na deficit = (desired Na – measured Na) * 0.6 * weight in Kg • Desired Na: 130 is ideal	**Fee fluid deficit** • 4mL/Kg for each 1 mEq if Na > 145 mEq • 3mL/Kg for each 1 mEq if Na > 170 mEq
			Fluid deficit containing electrolyte = Fluid deficit - Free fluid deficit
Step 3: Subtract the bolus if given			
Step 4: Divide total	Divide total over 24hrs.	Divide total over 24hrs.	Divide total over 48hrs. (The deficit should be corrected slowly over 48hrs.)
Step 5: Correct any on-going losses by mL-to-mL bases (منشوف وهو بالمستشفى إذا استفرغ مثلا ومنحسب قديش استفرغ وبناءا عليه منعطي زيادة سوائل)			

IV Fluids

Fluid	CHO (g/100ml)	Na (mEq/L)	K (mEq/L)
D_5W	5		
$D_{10}W$	10		
Normal Saline (NS)		154	
½ NS		77	
¼ NS		34	
3% NS		513	
Lactated Ringer's	0-10	130	4

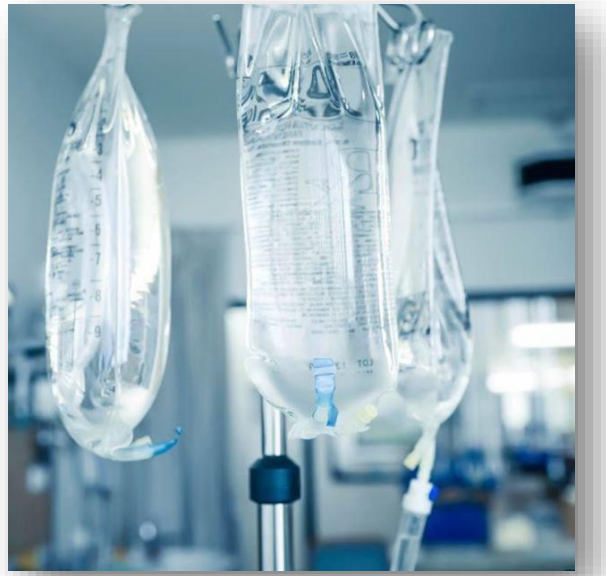
General Rules

- Isonatremic dehydration (corrected over 24hrs.)
  - Ideal is D5 ⅓ NS + 40 mEq/L KCl
  - But D5 ½ NS + 40 mEq/L KCl is safe to be used
- Hyponatremic dehydration (corrected over 24hrs.)
  - D5 ½ NS + 40 mEq/L KCl
- Hypernatremic dehydration (corrected over 48hrs.)
  - D5 ¼ NS + 40 mEq/L KCl

بالعربي اذا جوابك بعيد عن هذه الأرقام غالبا غلط !!
وبضع دائرة اختاروهم بدون ما تحسبوا بالغالب بطلعوا صح

Fluids

- A child 11 mothns old who weighs 10 kgs presents with moderate hyponatremic dehydration signs of dehydration. His Na+ level is 125



- **Calculate the sodium maintenance**
  - Maintenance = 2-4 =mEq/kg. So approximately 30 mEq
- **Calculate the sodium deficit**
  - Deficit = 100 mEq
  - Na correction = 60 mEq
  - Total = 100+60 = 160mEq

Scenario of child with head trauma and the following lab results

- **What's your diagnosis ?**
  - SIADH
- **Mention 2 lines of management:**
  - Fluid restriction
  - Hypertonic saline

- Na+: 110 mEq/L ,
- Urine output: 3ml/kg/hr

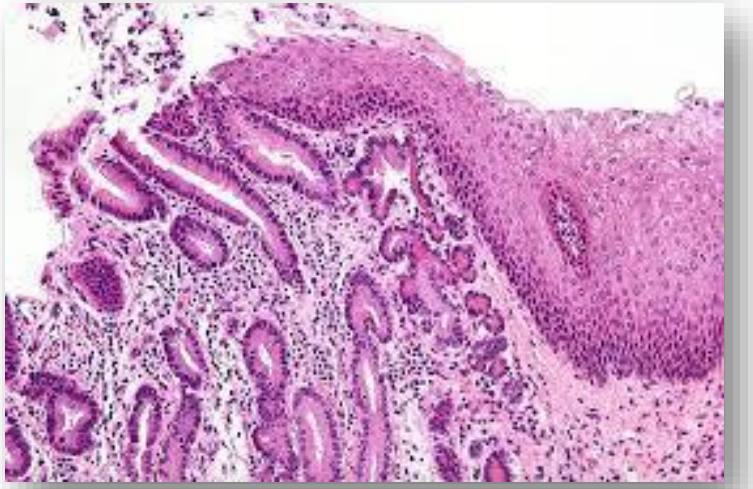
Gastrointestinal

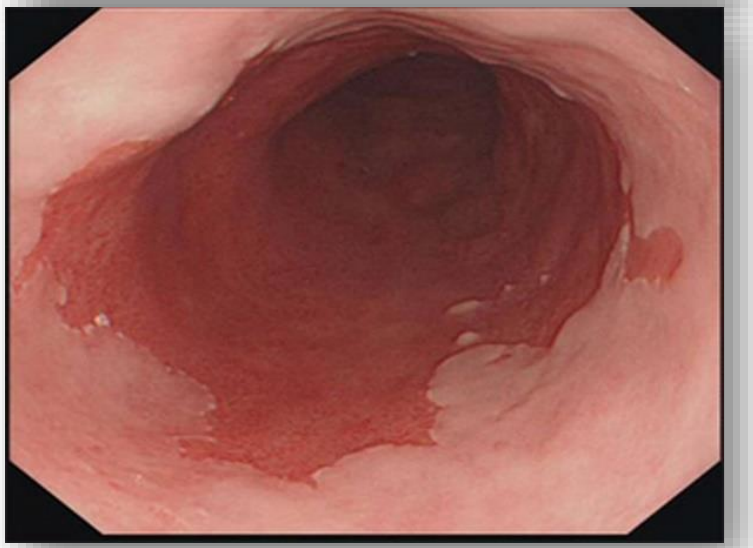
Gastrointestinal Lectures

Low-Yield

GERD

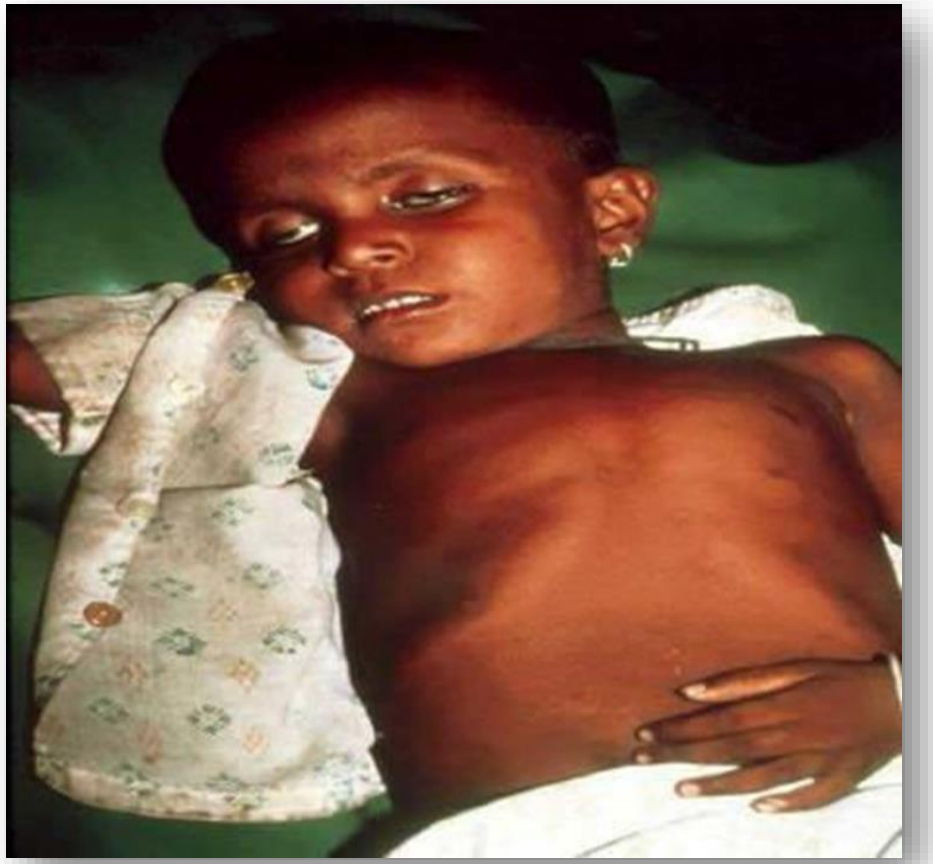
- **What is the diagnosis ?**
  - GERD
- **GI complication ?**
  - Stricture and esophagitis
- **Type of formula ?**
  - AR formula





A 7 years child with sever gastroenteritis

- **What's the most important sign seen in the picture?**
  - Sunken eyes



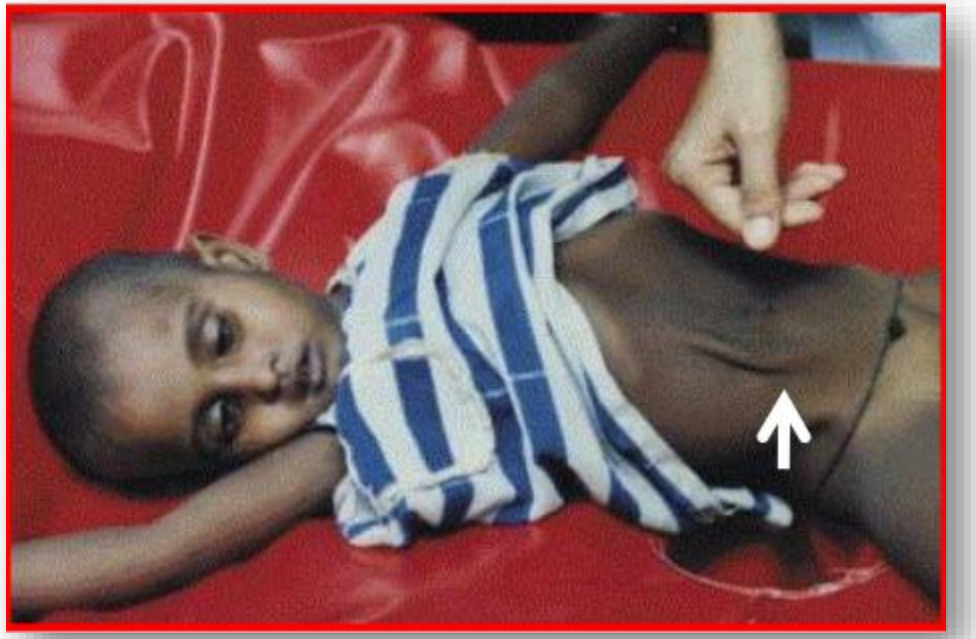
Dehydration

- The child came to the ER complain from vomiting and diarrhea for 3 days

- **What is the sign seen in the picture ?**
  - Decreased skin turgor

- **What is the degree of dehydration ?**
  - Severe dehydration

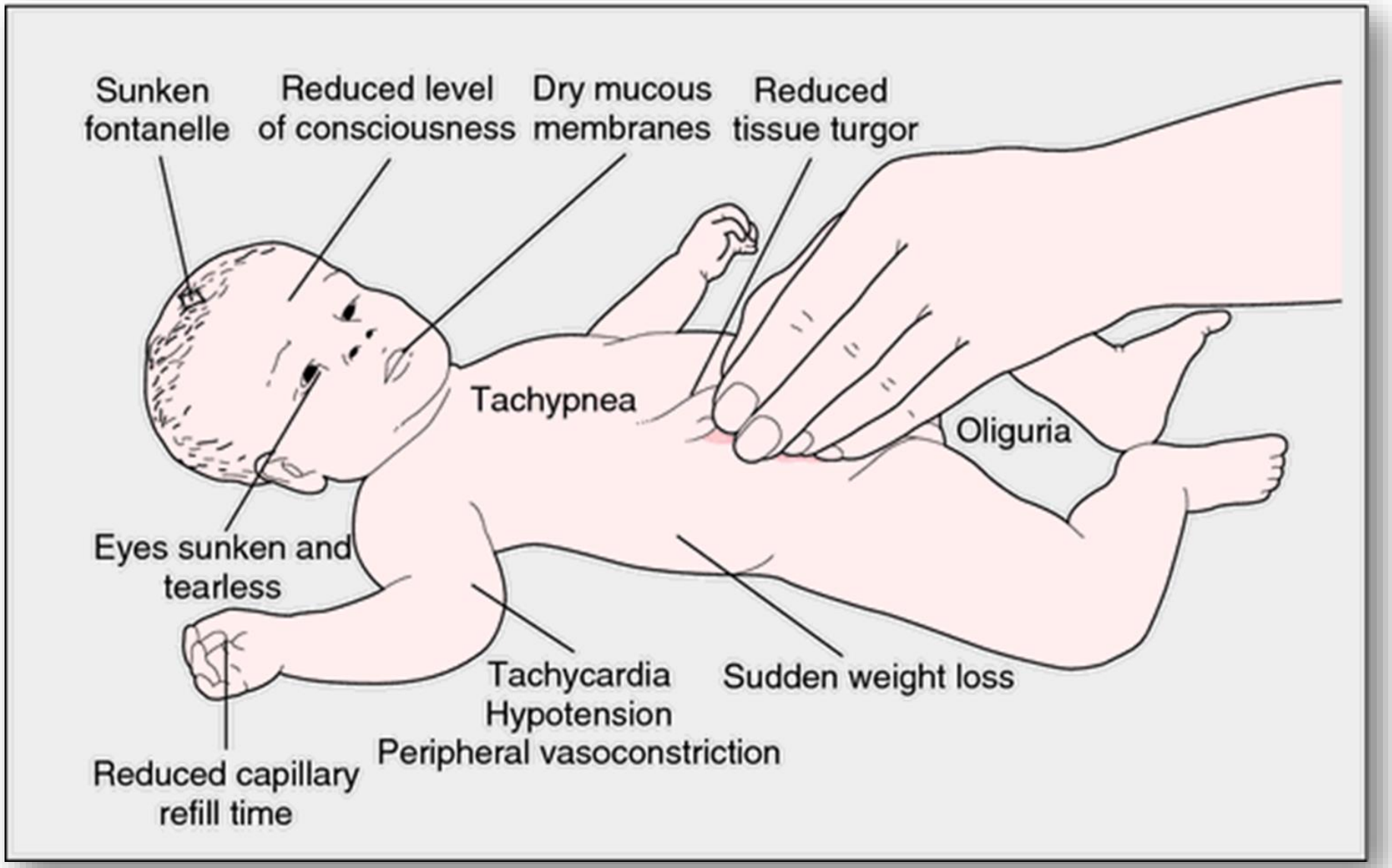
- **What is the first line in treatment with dose ?**
  - IV normal saline 0.9% 20 mg/kg as bolus



Signs and symptoms of dehydration

Symptom or Sign	No or Minimal	Mild to Moderate	Severe Dehydration
Mental status	Alert	Restless, irritable	Lethargic, unconscious
Thirst	Drinks normally	Drinks eagerly	Drinks poorly
Heart rate	Normal	Normal to increased	Tachycardia
Quality of pulses	Normal	Normal to decreased	Weak or unpalpable
Breathing	Normal	Normal or fast	Deep
Eyes	Normal	Slightly sunken	Deeply sunken
Tears	Present	Decreased	Absent
Mouth and tongue	Moist	Dry	Parched
Skin fold	Instant recoil	Recoil <2 seconds	Recoil >2 seconds
Capillary refill	Normal	Prolonged	Prolonged or minimal
Extremities	Warm	Cool	Cold, mottled, cyanotic
Urine output	Normal	Decreased	Minimal

Signs and symptoms of dehydration



Stool analysis

❖What is abnormal finding ?

- Presence of mucus and pus
- Presence of trophozoite and cyst (*E.Hystolitica*)
- Presence of RBCs
- Offensive odor

❖What is the treatment ?

- **Trophozoite**: metronidazole
- **Cyst**: luminal agent (iodoquinol or paromomycin)

Stool analysis

Physical

- **Color: Brown**
- **Odor: Offensive**
- **Consistency: Semi-formed**
- **Blood: N/L**
- **Mucus: +**

Microscopy

- **R.B.Cs: 3-5**
- **Yeast: N/L**
- **Starch: ++**
- **Vegetables: +++**
- **Fat: +++**
- **Protein: N/L**
- **Parasite: E.Histolatica**
- **Parasite Ova: N/L**
- **Parasites Cyst: E.Histolatica**

Stool analysis

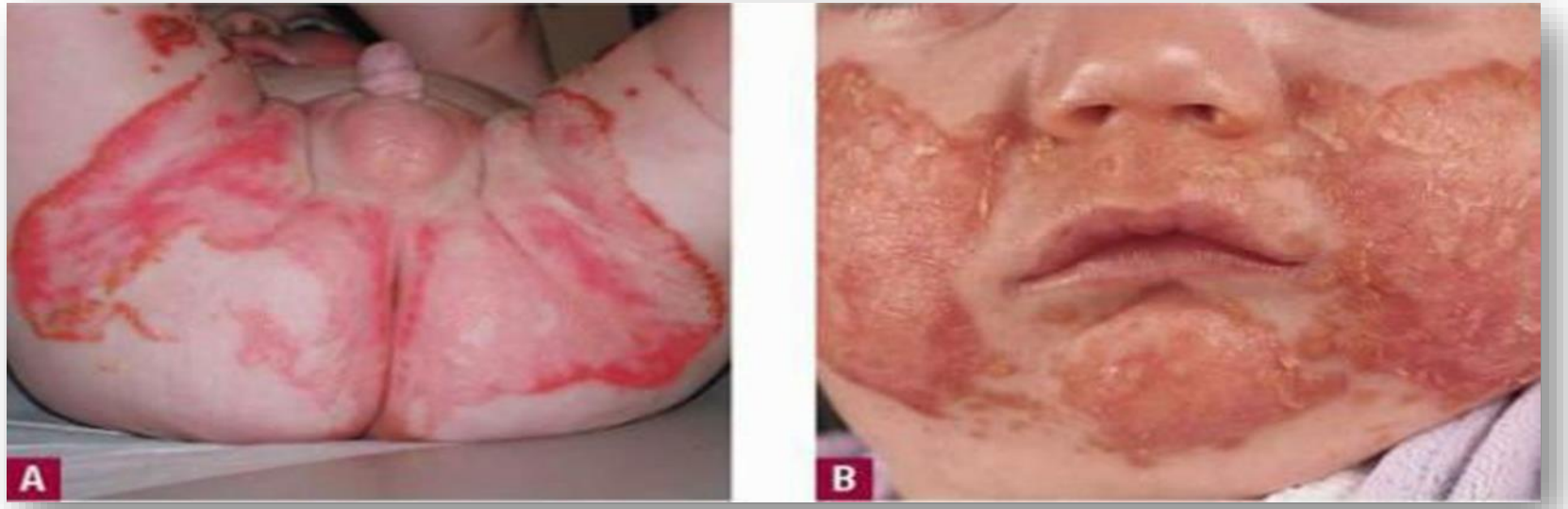
- Vomiting diarrhea seizure with stool test
- **What is the organism ?**
  - Shigella
- **What is the complication ?**
  - HUS
- **What is the treatment of choice ?**
  - Ceftriaxone

Stool analysis

- WBC – positive
- Mucus – positive
- Blood – positive
- No cysts or trophozoites

Malabsorption

- **What is your diagnosis ?**
  - Acrodermatitis enteropathica
- **What's the Most likely nutrient to be deficient?**
  - Zinc



Jaundice

- **Give 3 noninfectious causes of jaundice**
  - Galactosemia, tyrosinemia, a1-antitrpsin def
- **Give one drug that cause jaundice**
  - Acetaminophen

Presented with RUQ tenderness and elevated ALT

- **How can you confirm your diagnosis ?**
  - Serology (level IgM (active infection)/IgG (past infection)) or PCR of HAV-RNA from the stool or blood
- **How can you prevent his 4y old brother ?**
  - Vaccination with hygiene protocol (feco-oral transmission)
- **How can you prevent his 8-month-old brother ?**
  - Immunoglobulin Intramuscularly (IGIM) with hygiene protocol and vaccination
- **When to give this vaccine in Jordan ?**
  - 9 & 12 months

What is your Diagnosis for the following cases ?

1. Immunity secondary to vaccination
2. Acute HBV infection

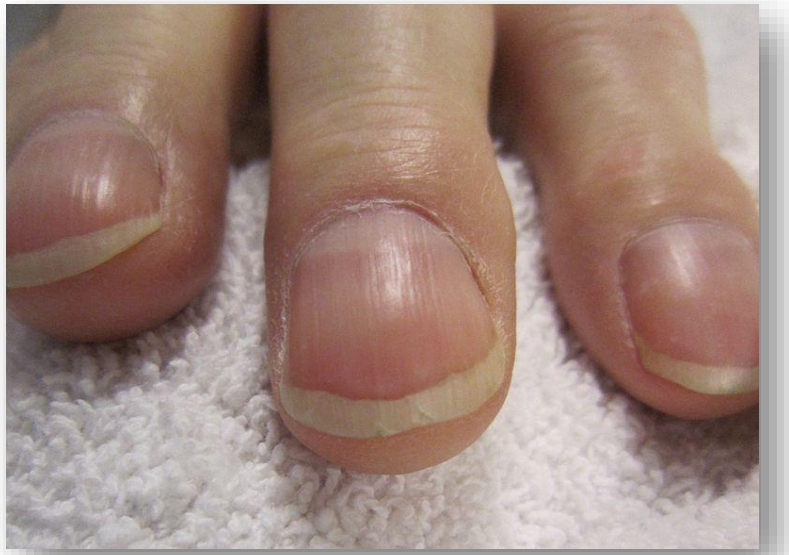
	HbsAg	HbsAb	HbcAb	IgM
1	Neg	Pos	Neg	Neg
2	Pos	Neg	Pos	Pos

Cystic fibrosis

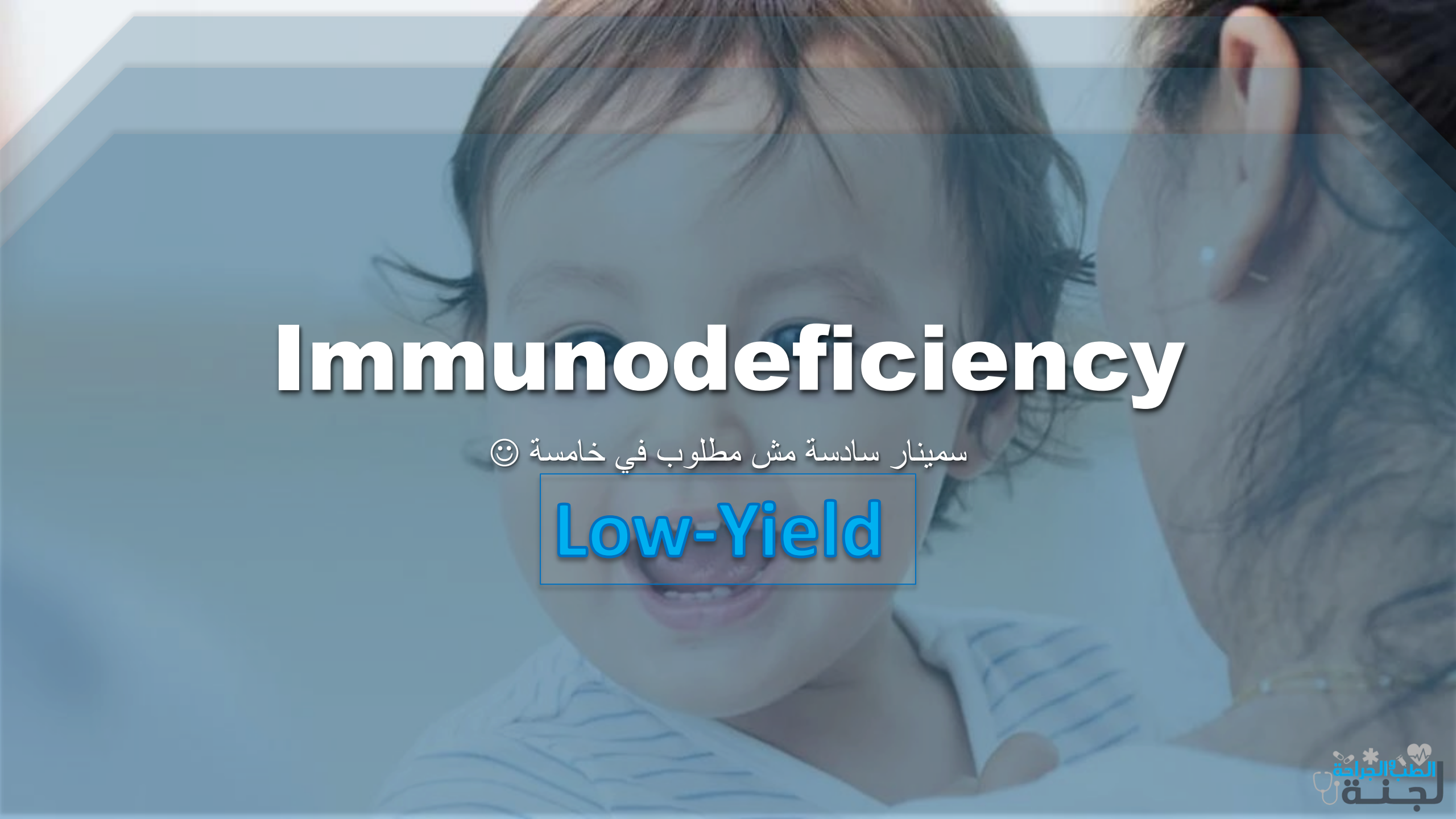
- Mention two diagnostic tests are useful to diagnose child with recurrent pneumonia and history of meconium ileus
  - Sweat chloride
  - Genetics

Cystic fibrosis

- **Name the sign**
  - Clubbing
- **Mention 2 physical finding in CF**
  - FTT and nasal polyps
- **Mention 2 investigation to confirm CF**
  - Sweat chloride test and genetics



6th Year Seminars



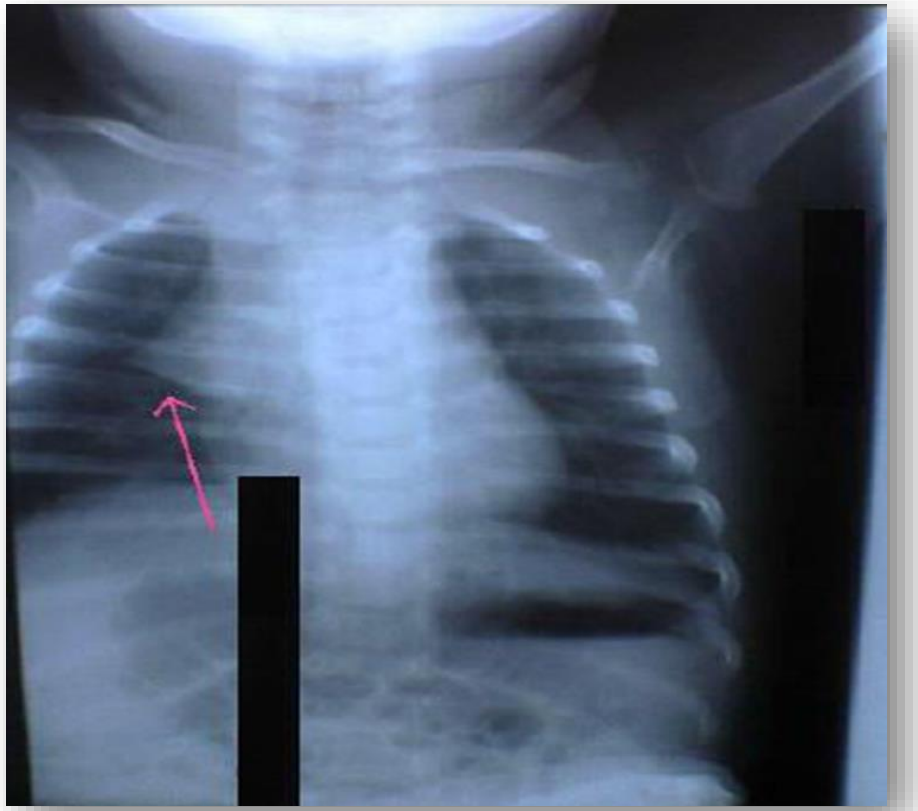
Immunodeficiency

سمينار سادسة مش مطلوب في خامسة ☺

Low-Yield

Thymus shadow

- **What's the name of this sign ?**
  - Sail Sign of thymus.
- **What's the most likely Dx. ?**
  - Normal X-ray.



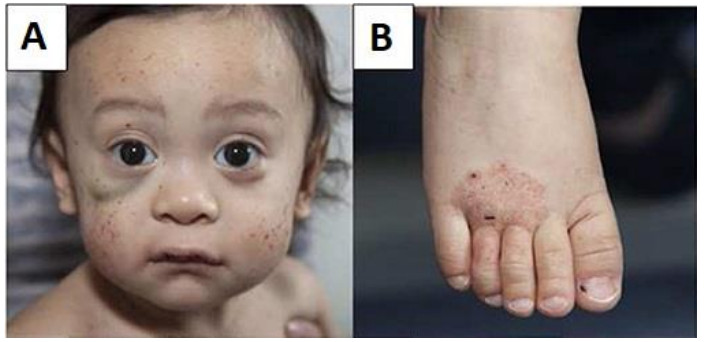
Immunodeficiencies

❖What neurological sign?

- Telangiectasia (ataxia telangiectasia)

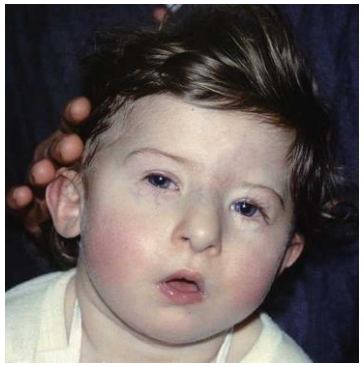
❖What expected CBC finding?

- Thrombocytopenia (Wescott Aldrich syndrome)



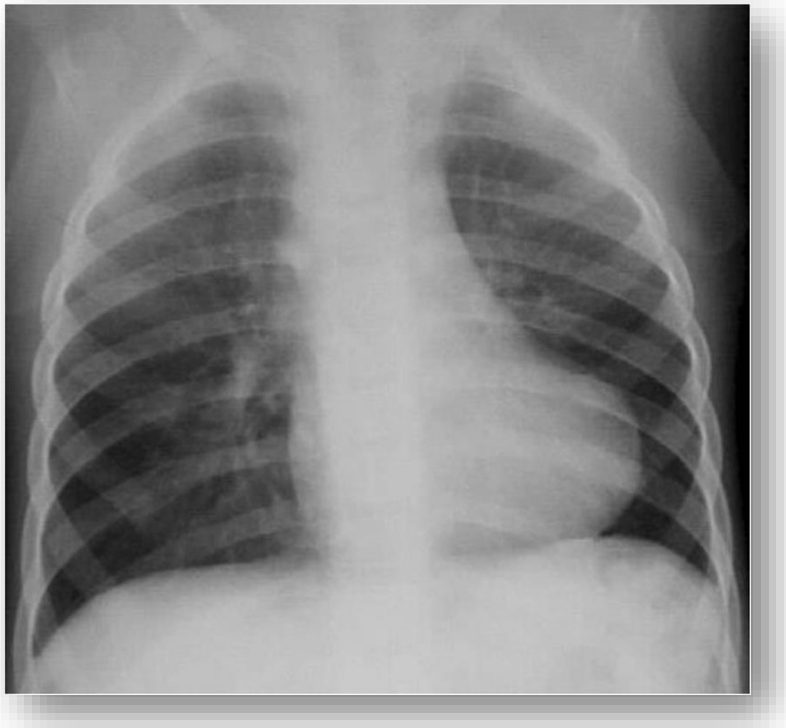
❖What electrolyte abnormality?

- Hypocalcemia (digeorge syndrome)

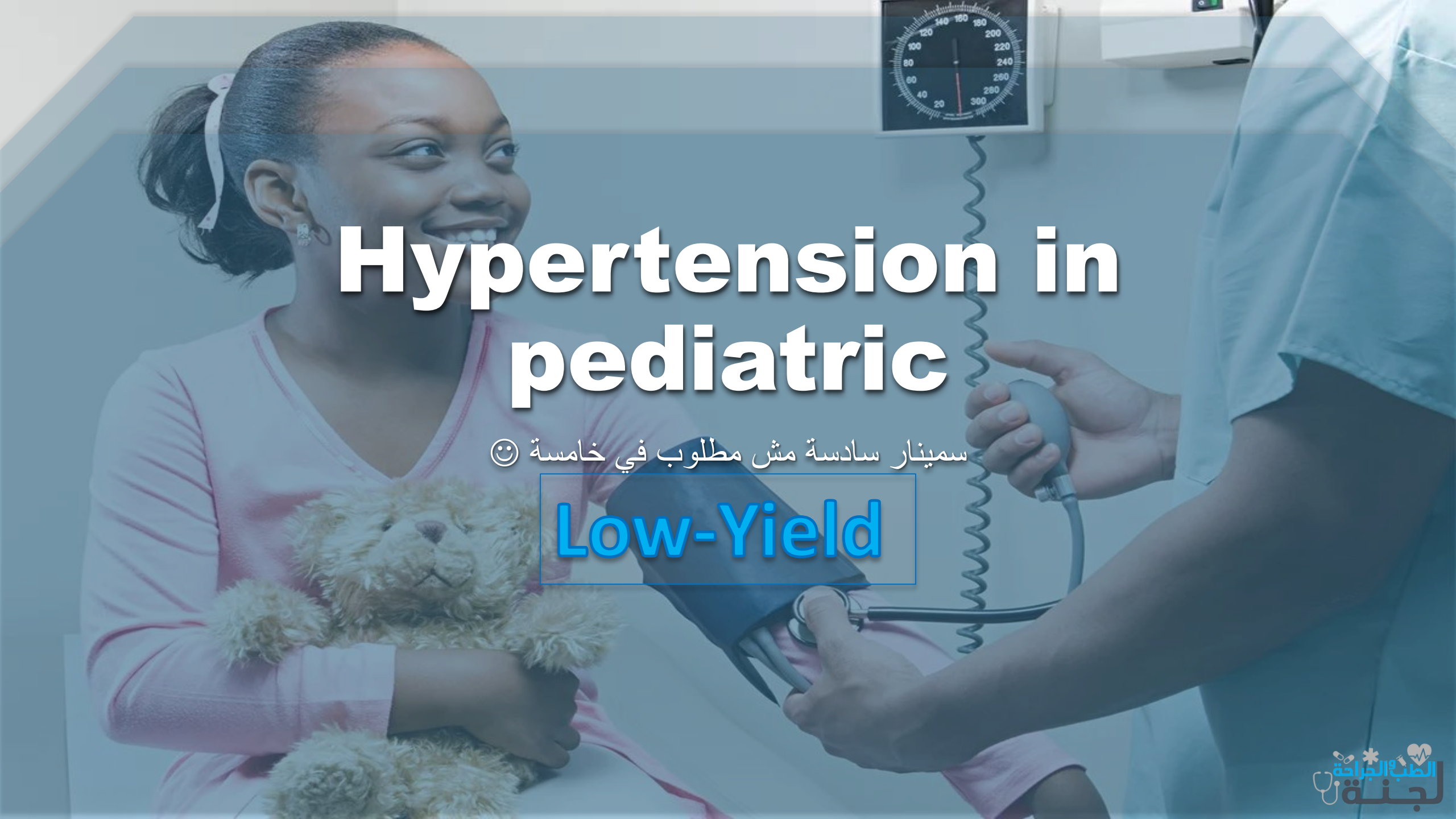


DiGeorge syndrome

- Scenario of DiGeorge syndrome features with X-ray picture of Boot-shaped heart



- **What are the findings in the X-ray ?**
  - Boot-shaped heart; TOF
- **How to treat TET spells ?**
  - Surgical intervention
- **What will cause his seizure ?**
- Hypocalcemia
- **What will cause His recurrent infections ?**
  - T-cell immunodeficiency



Hypertension in pediatric

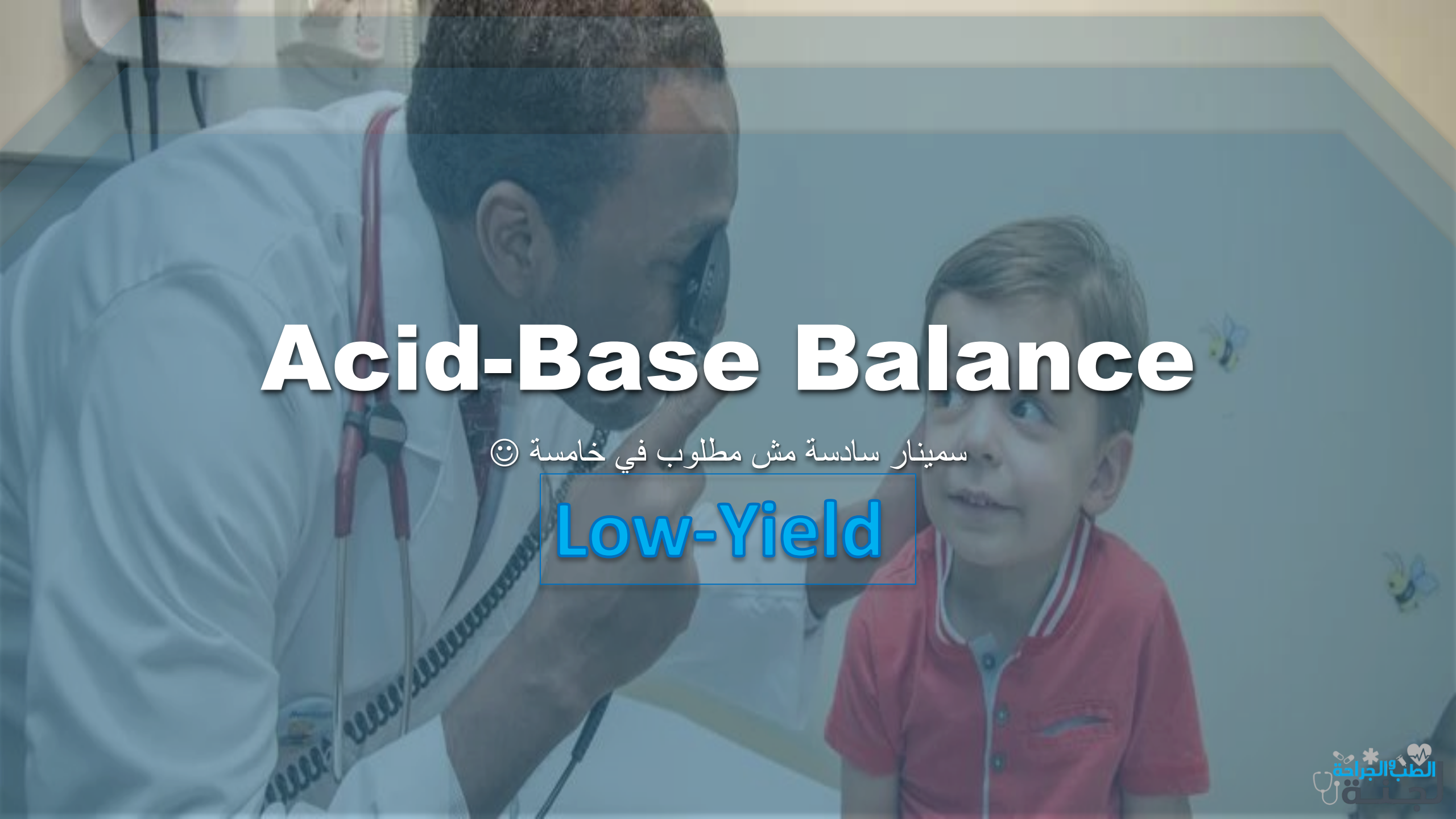
سمينار سادسة مش مطلوب في خامسة ☺

Low-Yield

4-year-old male, BP=116/70, and Hight =103 cm

- **Is this HTN and what stage ?**
  - Yes, stage 1
- **What is your drug therapy ?**
  - ACE inhibitors

3 years														
Height (in)	36.4	37.0	37.9	39.0	40.1	41.1	41.7	36.4	37.0	37.9	39.0	40.1	41.1	41.7
Height (cm)	92.5	93.9	96.3	99.0	101.8	104.3	105.8	92.5	93.9	96.3	99.0	101.8	104.3	105.8
50th	88	89	89	90	91	92	92	45	46	46	47	48	49	49
90th	101	102	102	103	104	105	105	58	58	59	59	60	61	61
95th	106	106	107	107	108	109	109	60	61	61	62	63	64	64
95th + 12 mmHg	118	118	119	119	120	121	121	72	73	73	74	75	76	76
4 years														
Height (in)	38.8	39.4	40.5	41.7	42.9	43.9	44.5	38.8	39.4	40.5	41.7	42.9	43.9	44.5
Height (cm)	98.5	100.2	102.9	105.9	108.9	111.5	113.2	98.5	100.2	102.9	105.9	108.9	111.5	113.2
50th	90	90	91	92	93	94	94	48	49	49	50	51	52	52
90th	102	103	104	105	105	106	107	60	61	62	62	63	64	64
95th	107	107	108	108	109	110	110	63	64	65	66	67	67	68
95th + 12 mmHg	119	119	120	120	121	122	122	75	76	77	78	79	79	80
5 years														
Height (in)	41.1	41.8	43.0	44.3	45.5	46.7	47.4	41.1	41.8	43.0	44.3	45.5	46.7	47.4
Height (cm)	104.4	106.2	109.1	112.4	115.7	118.6	120.3	104.4	106.2	109.1	112.4	115.7	118.6	120.3
50th	91	92	93	94	95	96	96	51	51	52	53	54	55	55
90th	103	104	105	106	107	108	108	63	64	65	65	66	67	67
95th	107	108	109	109	110	111	112	66	67	68	69	70	70	71
95th + 12 mmHg	119	120	121	121	122	123	124	78	79	80	81	82	82	83



Acid-Base Balance

سمينار سادسة مش مطلوب في خامسة ☺

Low-Yield

Metabolic acidosis

- **What is the metabolic disorder?**
  - Normal AG metabolic acidosis
- **Mention 2 Ddx ?**
  - RTA
  - Diarrhea

- pH = 7.2
- HCO3=11
- CO2=25
- Na=139
- Cl= 116

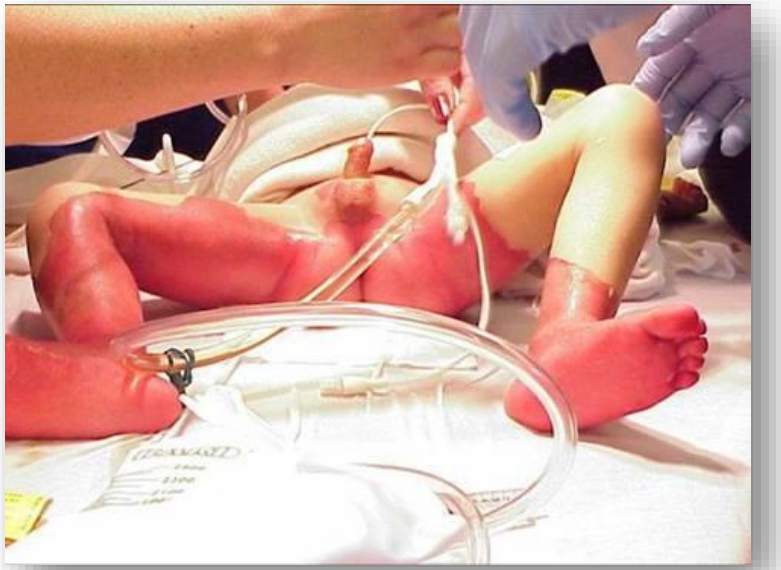
Child abuse

سمينار سادسة مش مطلوب في خامسة ☺

Low-Yield

6-month-old baby presented by lower limb bure

- **Two indication of abuse ?**
  - Burn in pre-mobile age and bilateral lower limb burn extending superiorly
- **Most common type of burn in child abuse ?**
  - Scald burn



Poisoning & Insect bites

سمينار سادسة مش مطلوب في خامسة ☺

Low-Yield

Paracetamol toxicity

- Paracetamol ingestion 125mg/5ml ingested 200ml (30 minutes)

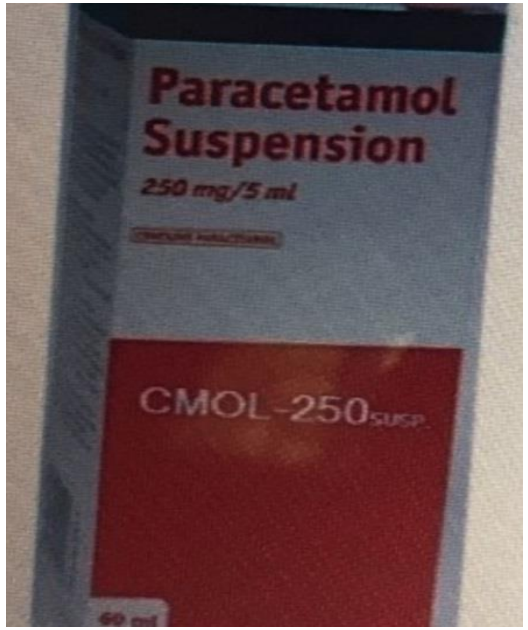
- **Is it a toxic dose and why ?**
  - Yes, it is above 150mg/kg (500mg/kg)
- **Mention 2 modalities of management ?**
  - Activated charcoal
  - gastric lavage or N-acetylcysteine??
- **Mention 2 lab investigations ?**
  - AST & ALT
  - INR & PTT
  - Paracetamol Serum level

Paracetamol toxicity

- 3 years old male patient whose weight is 15 kg, presented to the ER within one hour of taking 30 ml of the mentioned medication.

- Which of the following is correct regarding the approach?

a. Gastric lavage is the best method of decontamination
b. N-acetylcysteine should be given immediately
c. Paracetamol is highly beneficial before 4 hours
d. His dose is toxic for the patient
e. RUMAK-MATHEW chart is used for single ingestion cases



Bee Bite

- This patient has been admitted to ER after a bee bite, he was hypotensive.
- **What's your diagnosis ?**
  - Anaphylaxis
- **What's your immediate management ?**
  - Epinephrine injection