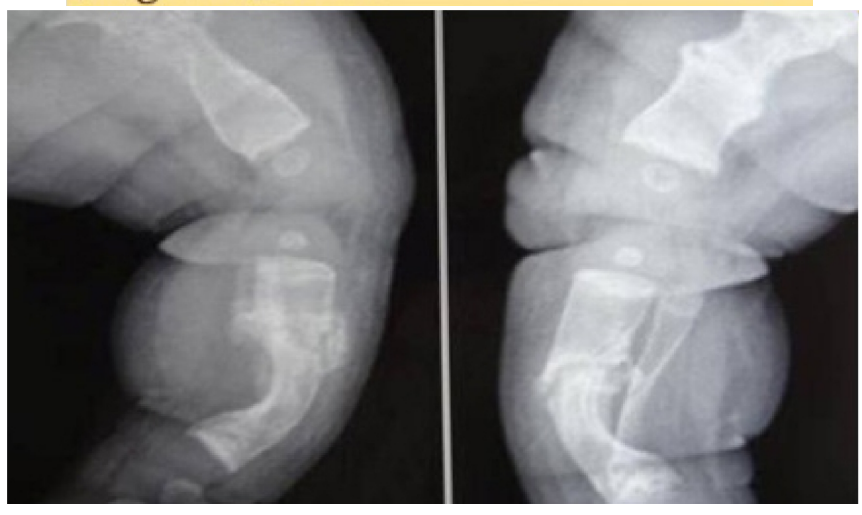
OSTEOGENESIS IMPERFECTA

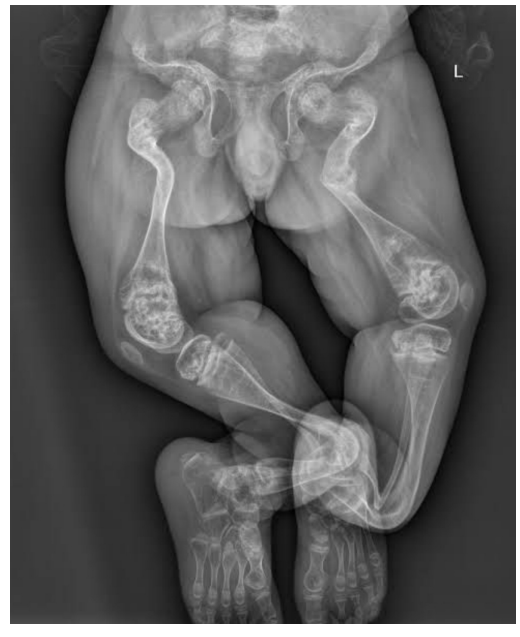
CASE

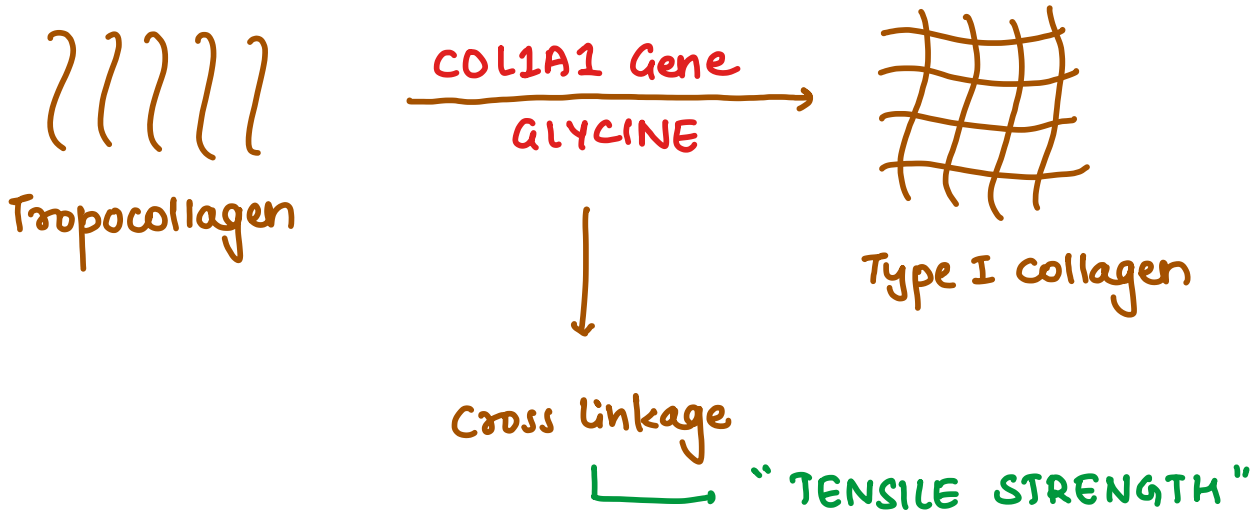
2 yr old baby
multiple fractures (thigh (femur), leg (tibia)) - pathological
No h/o trauma
on holding tight - petechial h'ages over the region
Blue eyes, Blue Sclera
Delayed Dentition.

Investigation :- x-Ray whole skeleton. (3'D's)
- 1) Diaphyseal #
- 2) Different stages of healing
- 3) Deformity

Osteogenesis Imperfecta / Brittle Bone Disease / Lobstein Vrolik Disease / Fragilitas Ossium







→ Rx : 2'B'
1) Braces
2) Bisphosphonates

COL 1A1 gene mutation

- GLYCINE SUBSTITUTION
- NO CROSS LINKAGE
- NO TENSILE STRENGH OF BONE

CLINICALLY preschool child

- MULTIPLE LONG BONE PATHOLOGICAL #S
- POOR & DELAYED DENTITION
- PETECHIAL HEMORRHAGES
- BLUE SCLERA (misnomer): bluish hue of uveal vessels through thinned out sclera

XRAY→3'DS

- Diaphyseal #s
- Different stages of Healing
- Deformities

Mx →2'Bs = Braces and Bisphosphonates

D/D = BATTERAD BABY SYNDROME

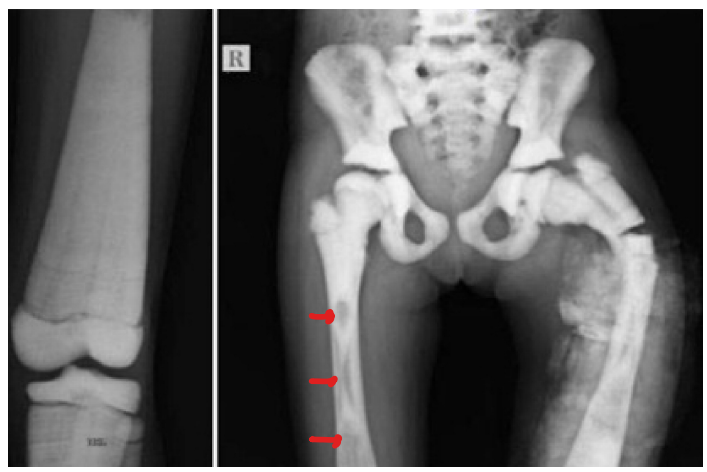
OSTEOPETROSIS

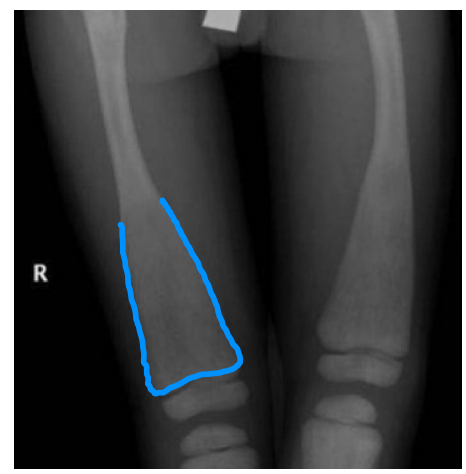
= Marble bone disease
= Albers-Schoenberg disease

CASE

ICU pt.
2 month old infant with bone marrow suppression
Pancytopenia, bleeding from multiple orifices
Septicemia, osteomyelitis
Hepatosplenomegaly

- x-Ray :-





2 'E's
↳ Endobones
↳ Erlenmayer flask Deformity

- Defect in carbonic anhydrase II proton pump leading to poor/defective osteoclastic bone resorption
  - Excess deposition of Ⓝ Osteoblastic bone formation
  - Thick/dense/sclerotic bone
  - Red bone marrow inside shaft of femur is replaced by bone

↓

BONE MARROW SUPPRESSION

- Pancytopenia
- Infectionsv
- Hepatosplenomegaly
- Bleeding episodes
- Osteomylitis of mandible

2 Es

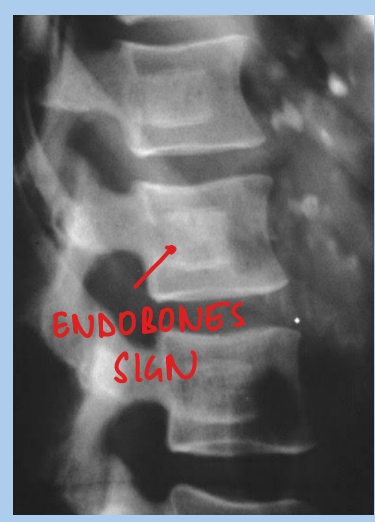
- Endobones sign (bone within Bone sign)
- Erlenmayer flask deformity

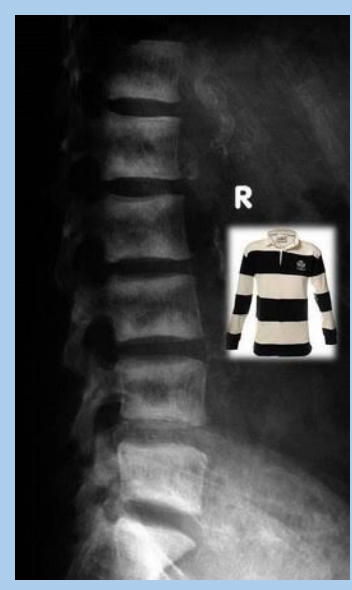
X-Ray

RUGGER JERSEY SPINE

Osteopetrosis < Renal osteodystrophy (ROD)

Mx: Bone marrow Transplantation (Poor prognosis)





RUGGER JERSEY SPINE
(R.O.D. > osteopetrosis)

PAGET'S DISEASE OF BONE

CASE 40 yr old ♂ , Asymptomatic

Had full body checkup, went to cardiologist & then referred to ortho.

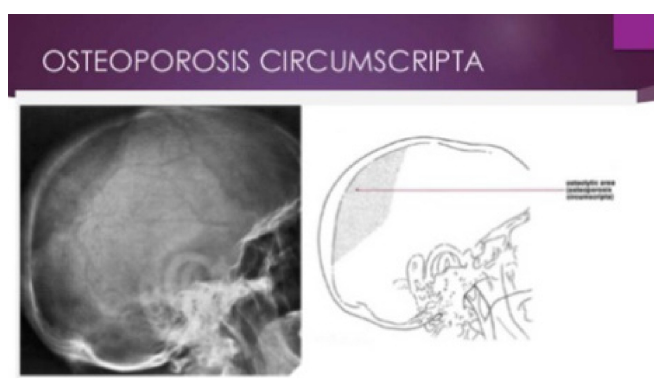
LFT : SGOT → Normal
SGPT → Normal
GGT → Normal
ALP → 1040 (normal : 40-140) ↑

USG Abdomen : Normal

- High Turnover bone disease
- Excess osteoblastic bone formation along with Excess osteoclastic Bone resorption
- Associated c- paramyxovial inf
- Asymptomatic pts. Initially
- 1st /earliest symptom: BACKPAIN
- MC site: PELVIS
- Cranial Nerve deficits (Sensorinevral hearing loss)
- High output cardiac Failure
- MC pre-malignant lesion for secondary osteosarcoma- (Incidence of conversion =1%)
- Biochemical HallmarK =8-10
  Times elevated levels of A.L.P.
- Bisphosphonates (D.O.C for Paget's ds of bone)
- Salmon calcitonin used as intranasal spray for pain relief in Paget's disease of bone

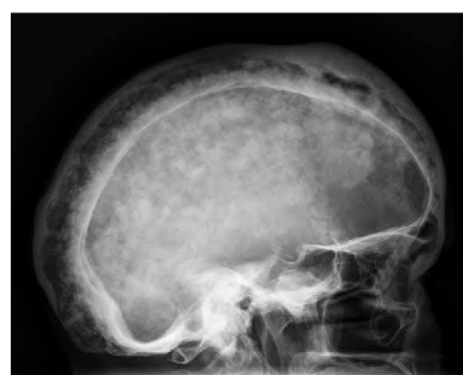
5 'P's → Pelvis (M/c Site)
Pain (backpain) (1st/earliest symptom)
Paramyxovirial infection (Associated infection)
Premalignant lesion for 2° osteosarcoma
Progressive SNHL (d/t compression of vestibulo-cochlear nerve)

X-RAY SIGNS IN PAGET'S DS. OF BONE

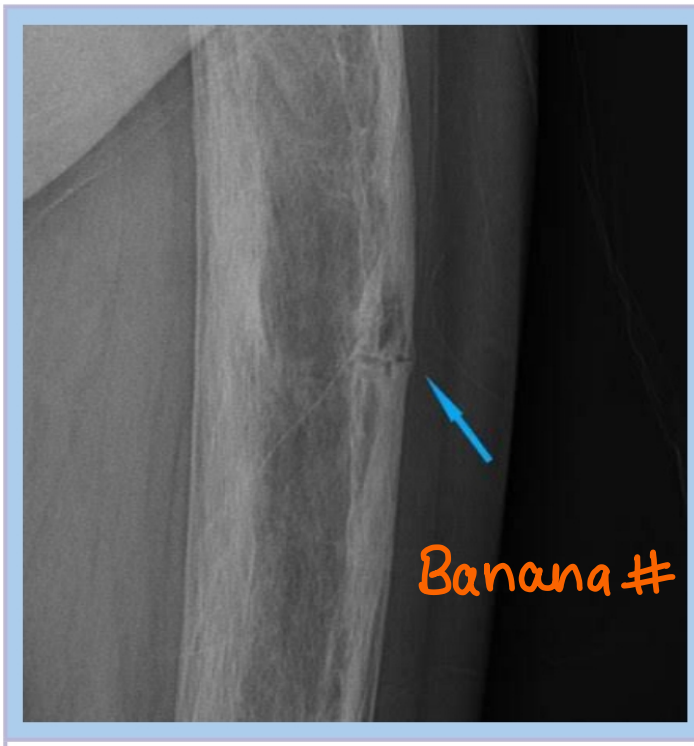


Well Circumscribed patch of osteoporosis

→ cotton wool skull



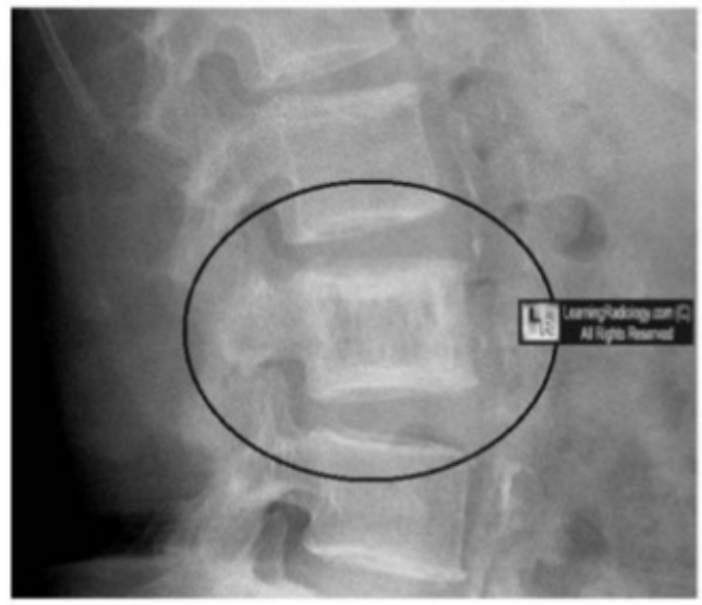
→ White fluffy deposits
→ Thick outer table of skull



X-Ray of Tibia (lateral tibia)

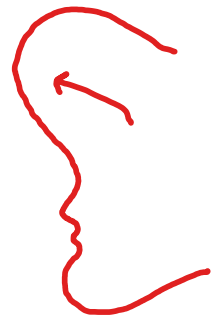
- Break in the anterior cortex in diaphyseal area

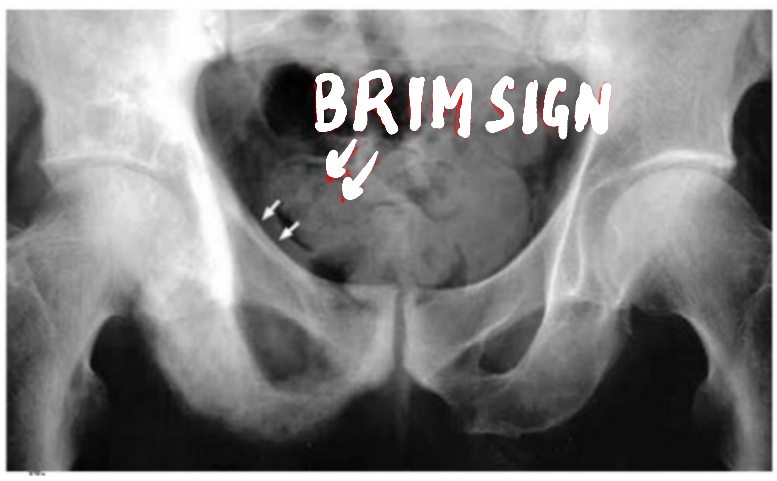
PICTURE FRAME VERTEBRA



All four end plates become dense

Excessively Protuberant frontal bone





BRIM SIGN

Thick & sclerotic

Iliopectineal line