Rickets

BY GROUP 24:

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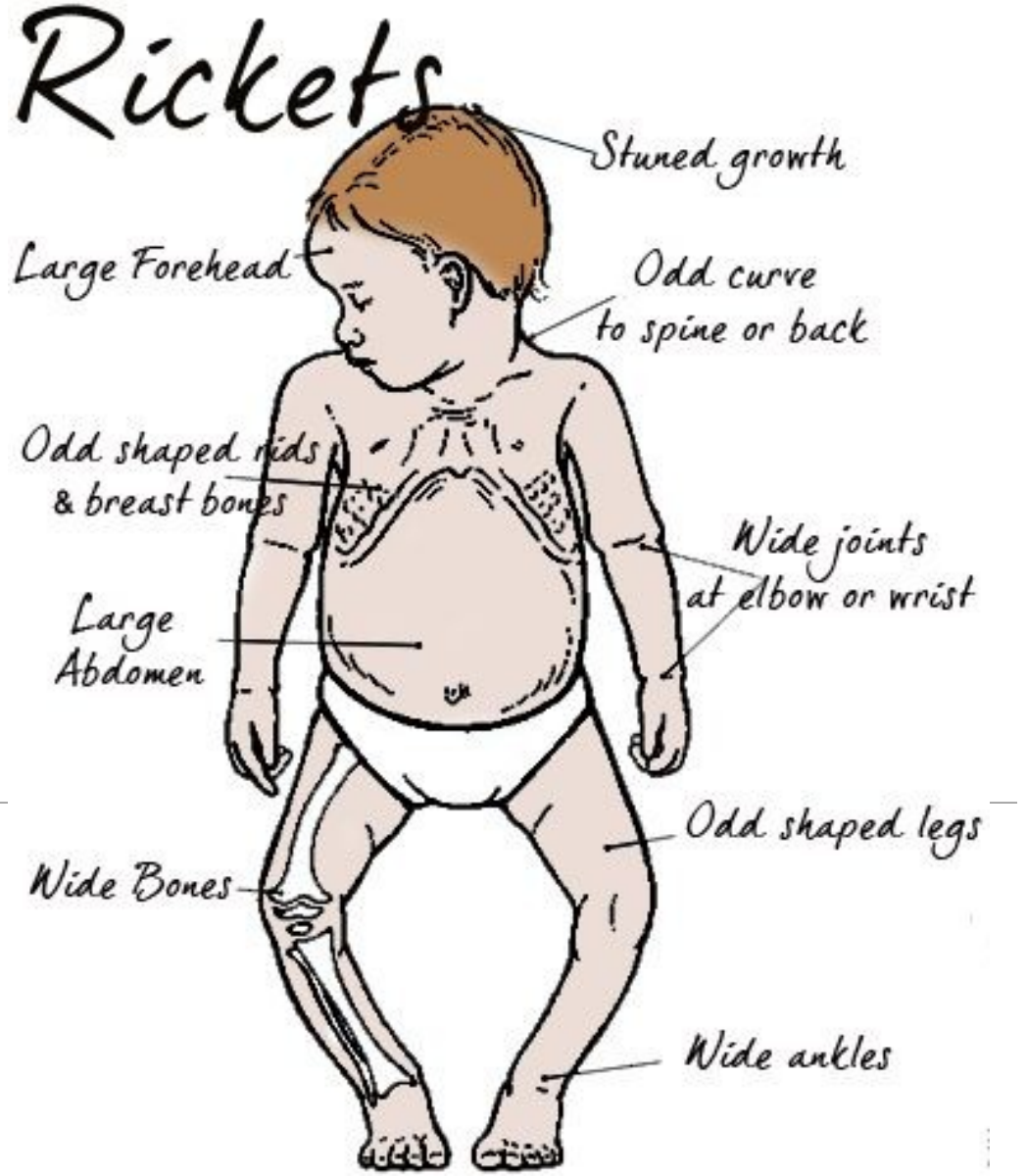


Outline

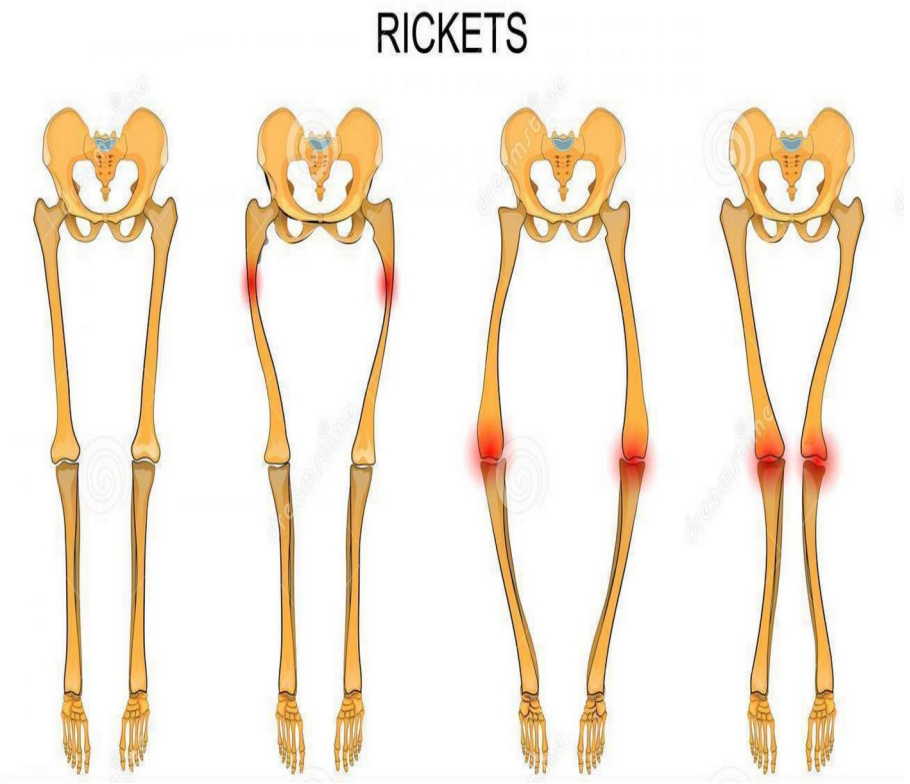
- Definition of Rickets
- Vitamin D, Vitamin D Activation and Physiological Action of Calcitriol (Active Form of Vitamin D)
- Pathogenesis of Infantile Rickets
- Types of Rickets
- Special Types of Rickets
- Hypophosphatemic Rickets
- Clinical Picture and Manifestation
- Diagnosis and Radiological Findings
- Complication of Infantile Rickets
- Treatment
- Prevention

Definition of Rickets

Metabolic disorder of infancy and childhood due to failure of mineralization of osteoid tissue of growing bone.

The most common cause of rickets is the lack of vitamin D and calcium in a child's diet which are essential for the development of healthy bones.

Rickets is most common in the first 2 years (between 6 months and 24 months) in which during this time the children experience rapid bone growth.

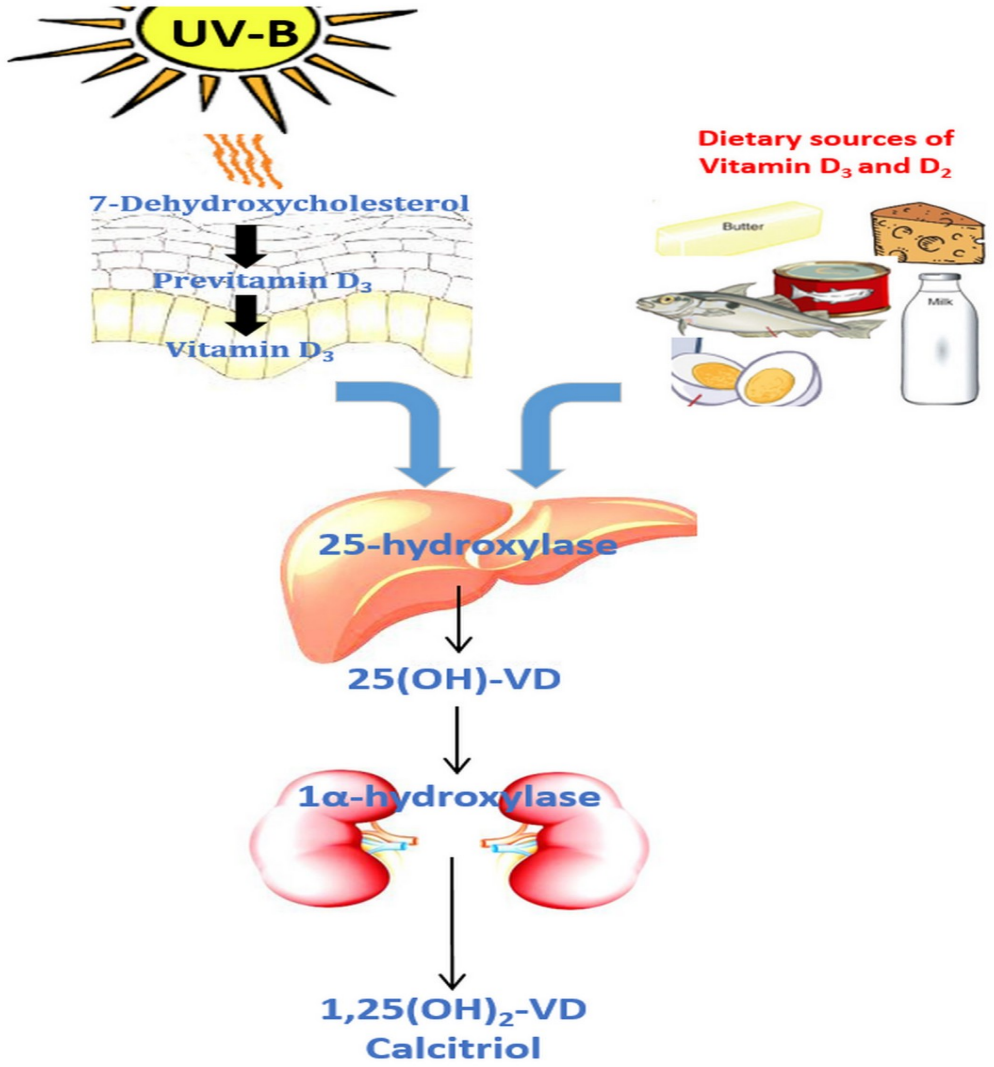


Vitamin D

- Fat soluble vitamin.
- Daily requirement
  - 400 IU/day (full term breast fed baby)
  - 800-1000 IU/day (preterm baby)
- 2 Natural Forms:
  - **Vitamin D3 (Cholecalciferol):** formed when the ultraviolet radiation of 7-dehydrocholesterol hits the skin, also present in food such as butter, egg yolk, fish and liver.
  - **Vitamin D2 (ergosterol)** is formed by the ultraviolet irradiation of ergosterol in green plants.

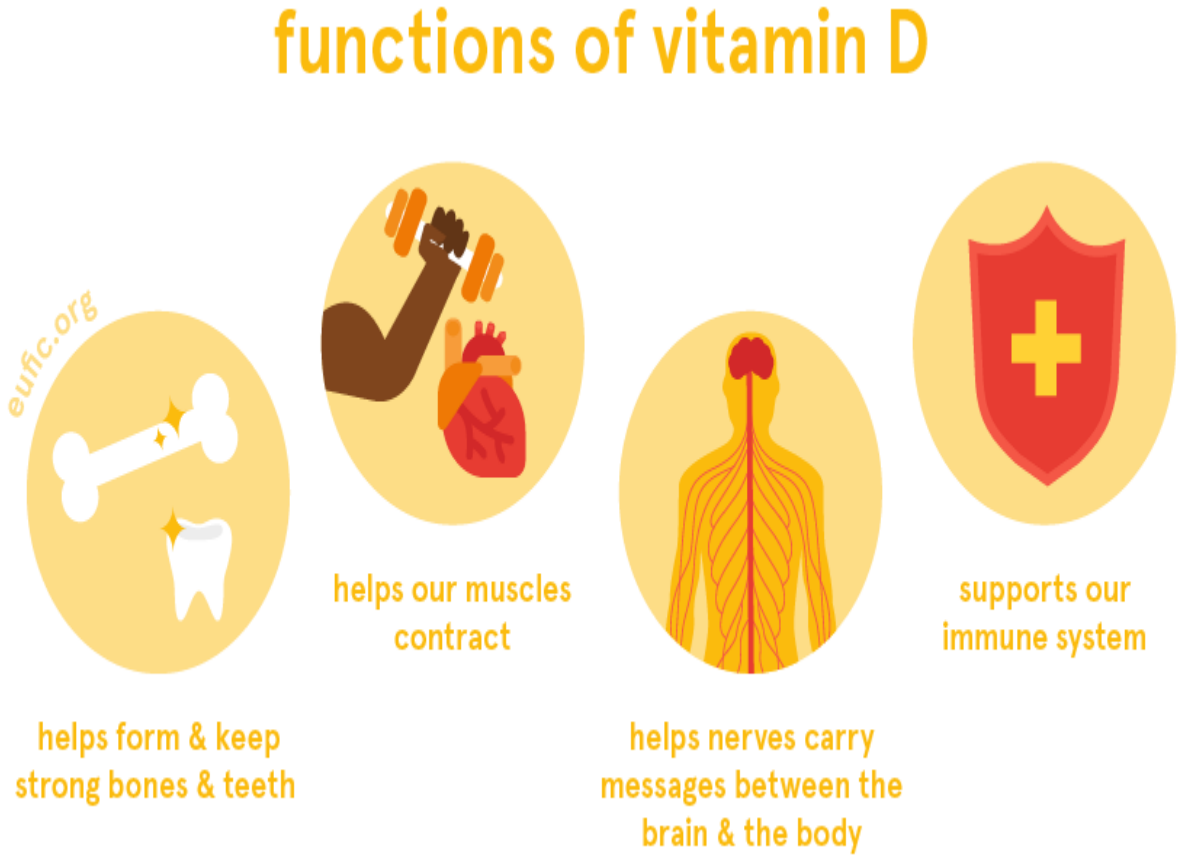
Vitamin D activa

- Vitamin D3 and D2 are biologically inactive forms.
- Vitamin D3 and D2 are transported to the liver to be hydroxylated to 25-hydroxyl Vit D (calcidiol) (not active form).
- Then in the kidney hydroxylated to 1,25 dihydroxy Vit D (calcitriol) = BIOLOGICALLY ACTIVE FORM



Physiological action of Calcitriol (active form of Vitamin D)

- GIT: promotes Calcium and Phosphate absorption in the small intestine.
- Kidney: Increase calcium and phosphate reabsorption
- Bone and teeth: affects calcium and phosphate resorption from old bone and teeth and deposition them in the newly formed bone.
- Immune systems: helps with the differentiation of immune cell preventing infection.

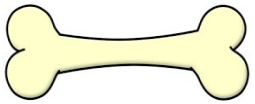


Pathogenesis of Infantile Rickets

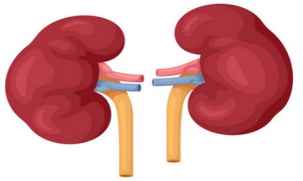
Vitamin D Deficiency

Decrease intestinal absorption of Ca and Ph

Decrease Ca level in blood and ECF



Stimulate of parathyroid gland (PTH)



Bones: increase mobilization of Ca

Normal Serum Ca level (9mg/dl)

Kidneys: decrease reabsorption of Ph

Decrease Serum Ph level (<3mg/dl)

Low Ca X P solubility product (<30)

Failure of calcification of intercellular matrix

In Cartilage: Mature cartilage cells will not die

In Zone of Provisional Calcification: excess non calcified osteoid tissue

Types of Rickets

1. Nutritional Rickets

- Prolonged exclusive breastfeeding without vitamin D supplement, breast milk lack vitamin D
- Lack of sun exposure
- Preterm baby, twins, black baby
- Malabsorption: celiac disease, cystic fibrosis
- Rachitogenic diet (carbohydrates, cereal, cow milk)
- Children on anticonvulsant drugs such as phenytoin and phenobartitone.

2. Non-nutritional Rickets

- Renal Rickets
  - Chronic kidney disease
  - Renal tubular acidosis
  - Fanconi syndrome
  - Familial hypophosphate rickets
  - Vitamin dependent Rickets Types 1
- Hepatic Rickets
  - Liver cirrhosis
  - Biliary atresia
- Other :Vitamin dependent Rickets Types 2

Special Types of Rickets

	Vitamin D Dependent Rickets Type I	Vitamin D Dependent Rickets Type II
Mode of inheritance	Autosomal Recessive	Autosomal Recessive
Defect	Decrease in 1 alpha hydroxylase enzyme	End organ resistance to vitamin D
Onset	Early 3-12 months	Early 3-12 months
Clinical picture	As nutritional Rickets	As nutritional Rickets
Associated with		Short stature and alopecia
Level of 1,25 dihydroxy-cholecalciferol	Low	Normal
Treatment	Vitamin D (calcitriol) + Oral calcium	Vitamin D (calcitriol) high dose + oral calcium

Hypophosphatemic Rickets

- X-linked dominant disorder (XLD)
- Characterized by decrease in renal tubular reabsorption of phosphate → loss of phosphate in urine (phosphaturia)
- Common in girls, sever in boys
- Onset: usually after the age of 1 year
- The commonest presentations:
  - Similar to nutritional rickets but sever deformity
  - Growth retardation (short stature)
  - Bow legs
  - Rickets unresponsive to the usual dose of vitamin D
- Investigation:
  - Serum calcium normal or low
  - Phosphate: low
  - ALP: High
  - PTH: normal

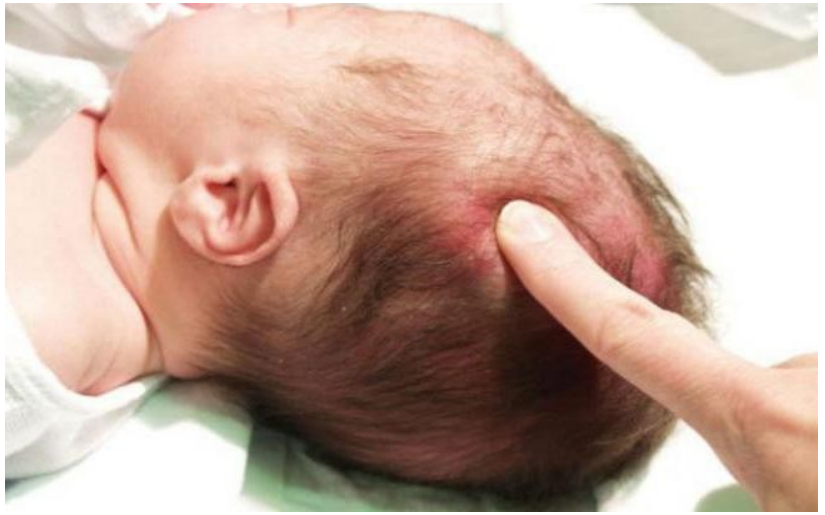
Treatment: Calcitriol and phosphate (oral)

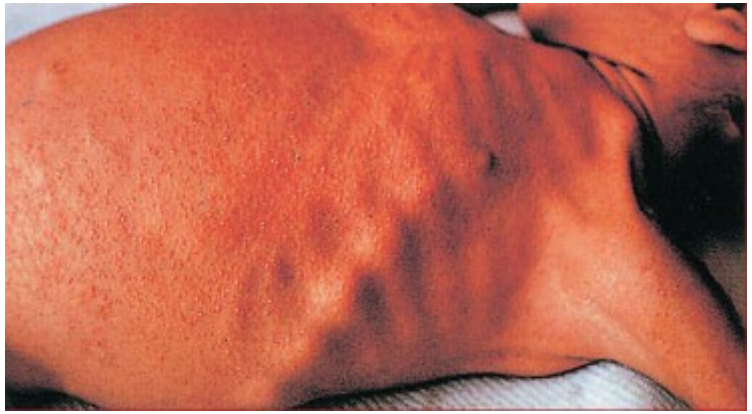
Clinical Picture (Early Manifestation)

1. Anorexia, irritability, increased sweating

2. **Craniotabes** (thinning of the outer table of the skull) is the earliest bony changes, pressure on the skull giving a ping pong sensation

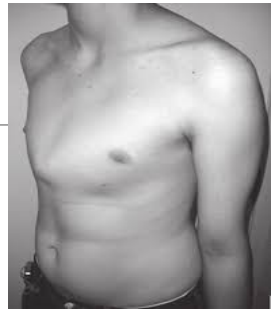
3. **Rachitic rosary:** prominent enlargement of the costochondral junctions seen and felt as arrow of beads.

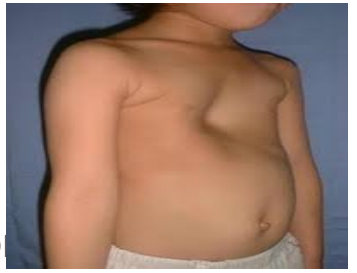


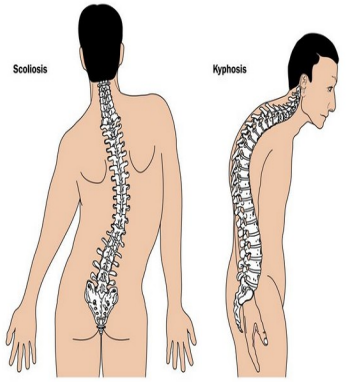


Advanced Rickets manifestnation

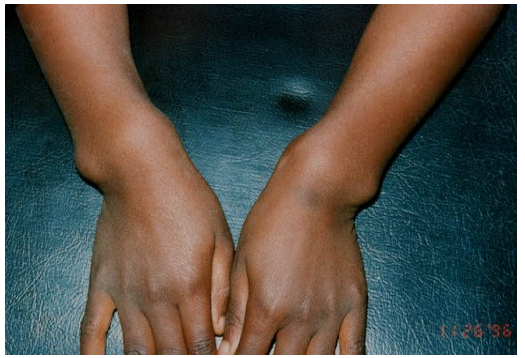
- Symptoms
  - Delayed sitting and standing
  - Delayed teeth eruption
- Signs
  - Head
    - Craniotabes: usually disappear by the end of 1st year
    - Wide anterior fontanelle and delayed closure
    - Frontal posing and square shaped head
    - Size—large (macrocephaly)
    - Delayed eruption of teeth
- (Continuation of Signs)
- Thorax
  - Ricketic rosary
  - Chest deformity: pigeon shaped chest, funnel shaped chest
  - Harrison Sulcus: It's transverse groove at insertio of diaghram
  - Spinal kyphosis or scoliosis
- Muscle: hypotonia
- Neurological: Tetany due to hypocalcemia (convulsion, stridor, carpopedal spasm)

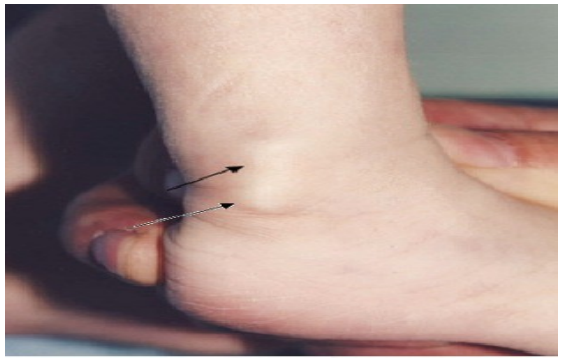


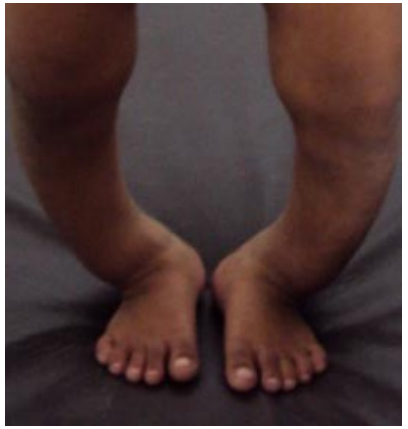


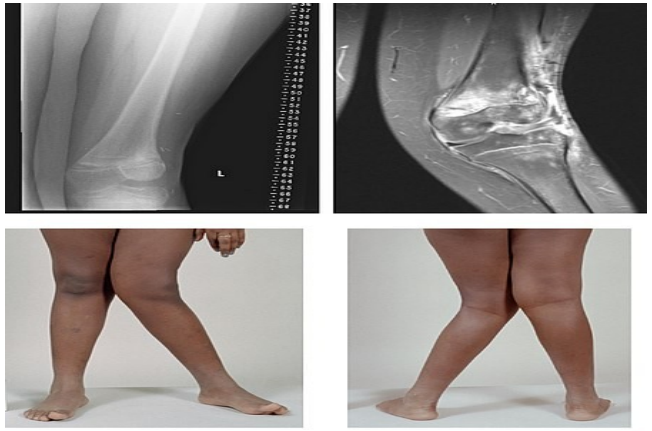


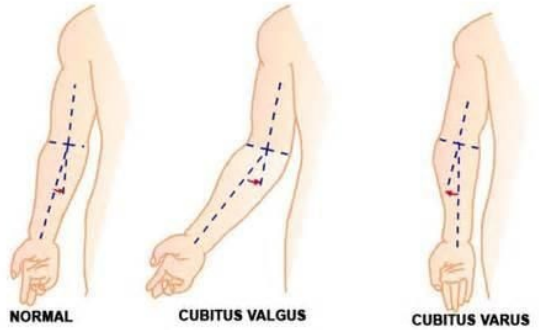
- Extremities
  - Widening of wrist and ankle joints
  - Marfan sign: transverse groove at medial malleolus
  - Deformity:
    - If the infant is crawling: cubitus varus or valgus
    - If infant walking: genu varum (bow legs) or genu valgus (knock knee)



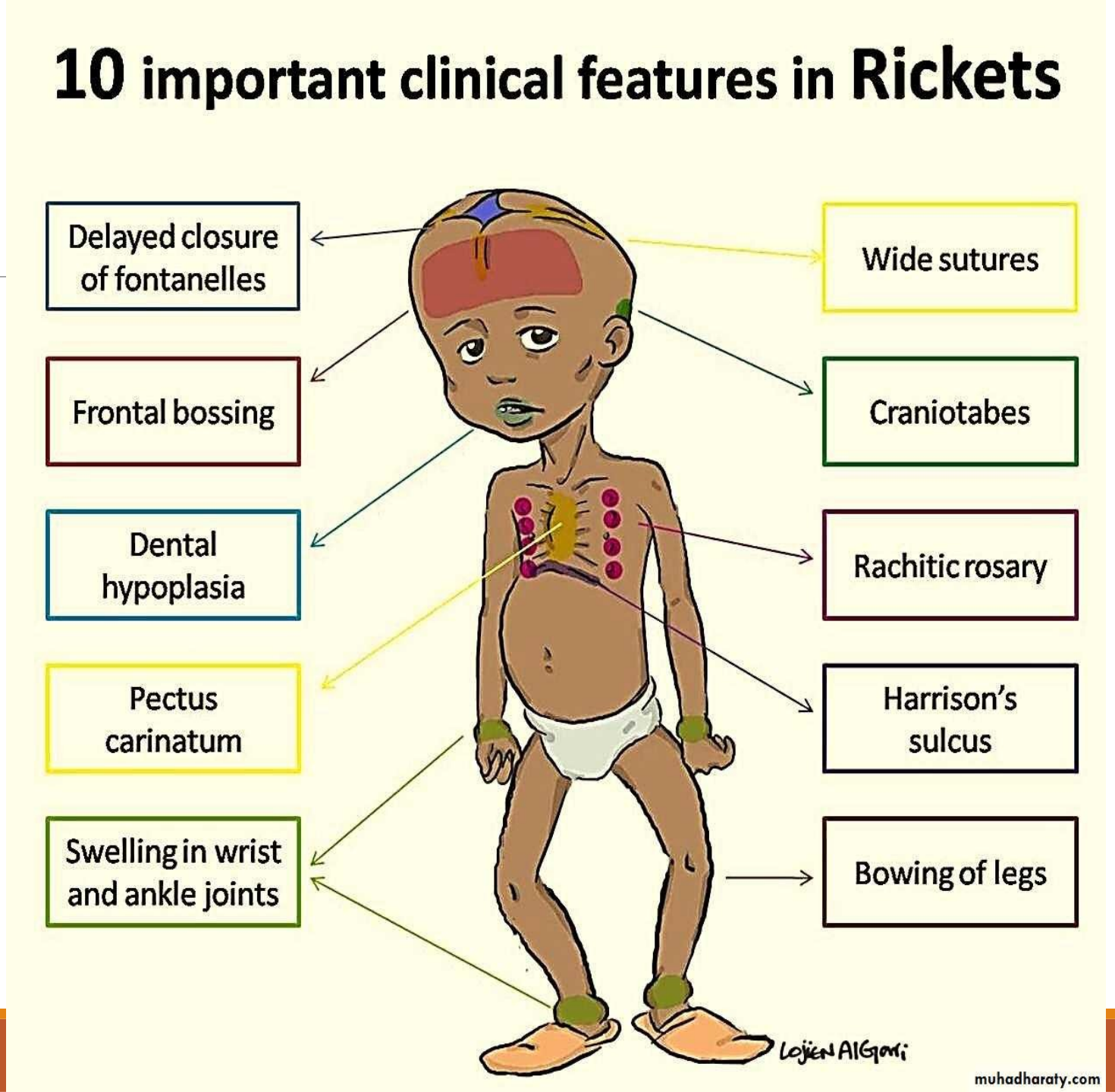








Clinical Picture Rickets

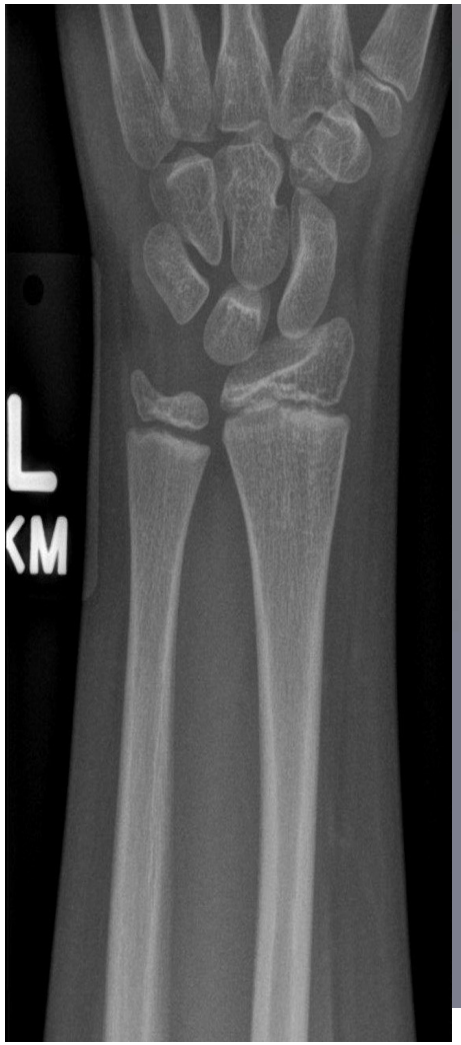


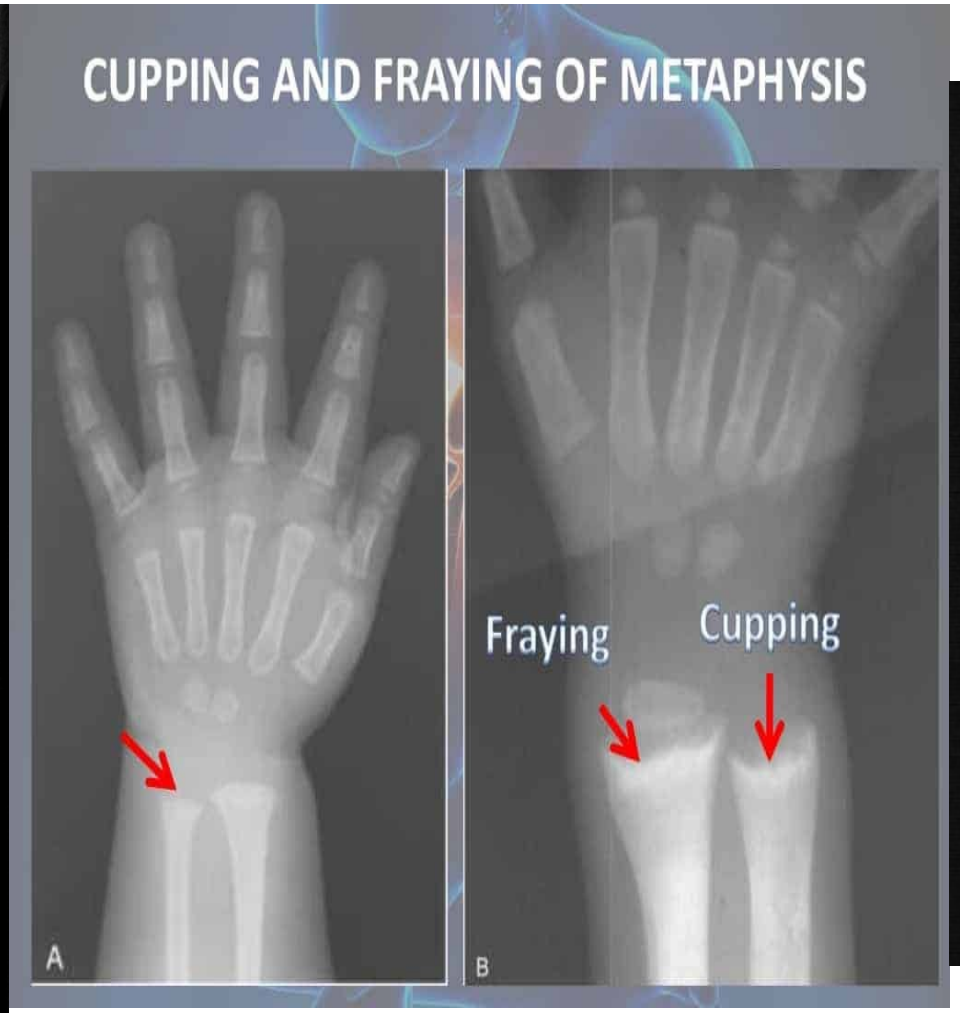
Diagnosis of Rickets

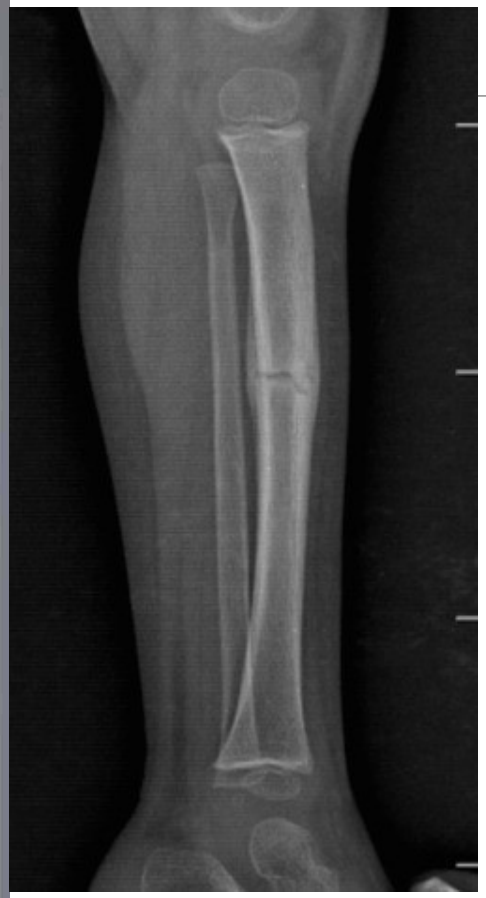
Biochemical (Laboratory) findings:

- Serum Ca: normal due to compensatory hyperparathyroidism or low
- Serum phosphate: low due to compensatory hyperparathyroidism—Renal Rickets (High)
- Calcium X phosphate solubility product is low <30 (N >45)
- Serum Alkaline phosphatase ALP: increased (most sensitive) (not specific) (1^{st})
- Serum cacidiol and serum calcitriol: low
- Serum vitamin D level: low
- PTH level: High

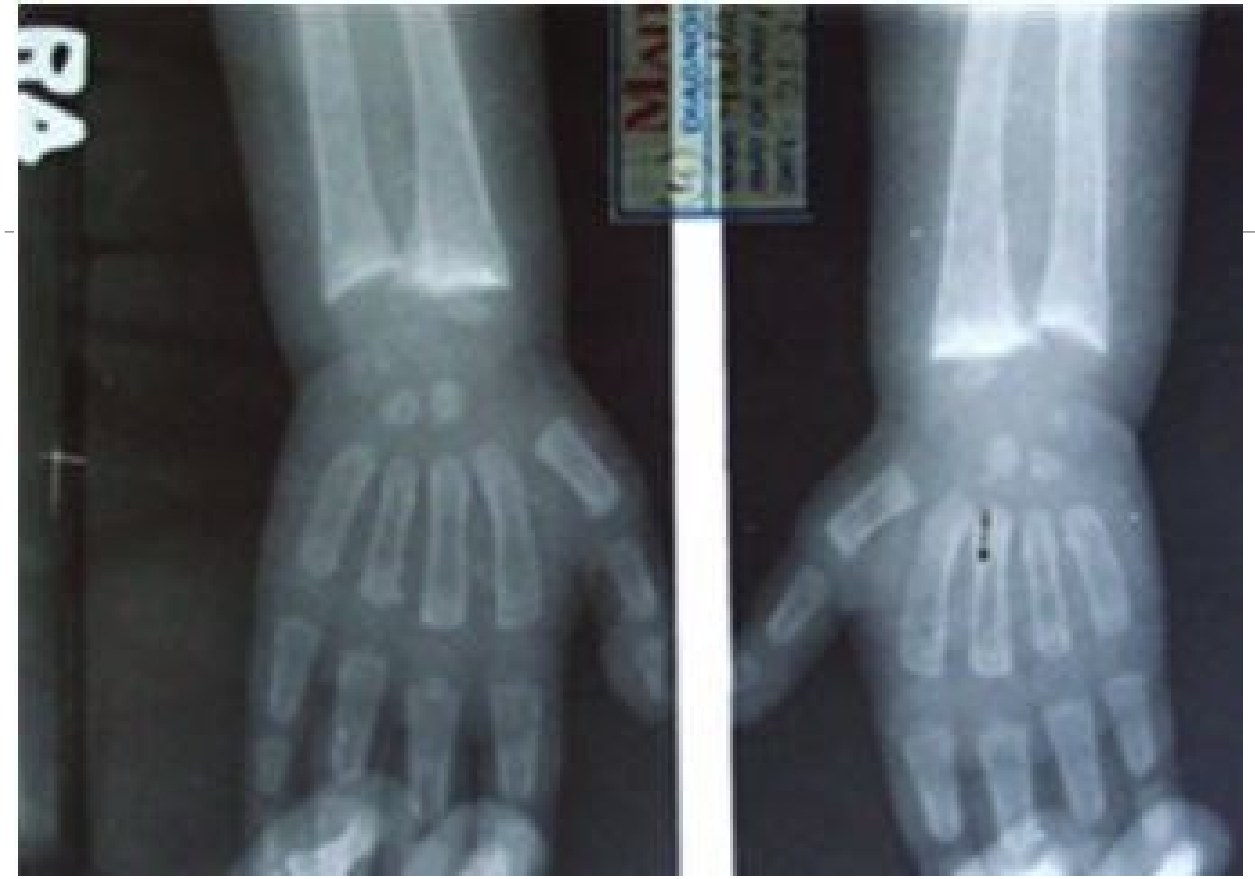
Radiological findings:







1-Active rickets :
1-cupping
2-fraying
(irregular end of joint)
3-wide joint space
4-decrease bone density
5-greenstick fracture



2-Healing rickets:
After calcium being given, evidence of healing may take from 2 to 3 weeks to appear on x-ray.
*Zone of preparatory calcification : Its appearance indicates initial healing.
It's an irregular band of calcification separated from ends of long bone by the uncalcified metaphysical zone of osteoid tissue.

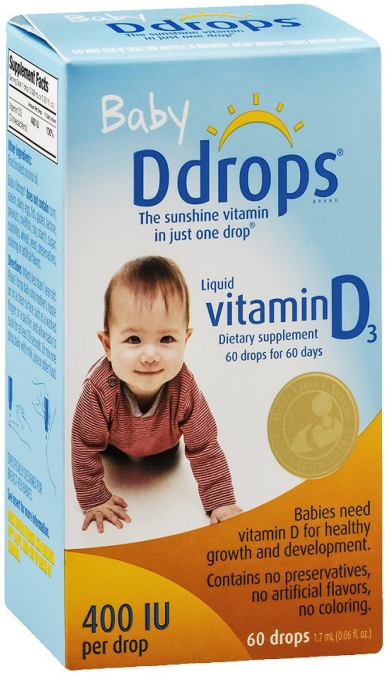
3-Healed Rickets: After about one month of therapy (vitamin D injection) no sign of active rickets.

Complication of Infantile Rickets

- Respiratory: chest deformities leading to:
  - Recurrent chest infection
  - Pulmonary atelectasis
- Neurological: hypocalcemic tetany
- Anemia: Iron deficiency anemia (common association)
- Skeletal: fractures and sever bone deformity

Treatment of Rickets

- Daily oral dose of 1000-4000 IU (2-4 weeks) or single huge dose 600,000 IU intramuscular (IM)
- After healing complete reduce dose to 400 IU/day as maintance dose
- Ca Supplement (Ca gluconate) in sever cases or with tetany
- Treat the complication
- Advice: Exposure to sun, start weaning, vitamin D prophylaxis: 400 IU/day orally



Prevention of Rickets

- Exposure to ultraviolet rays (direct sunlight, clear sky)
- Diet rich in Vitamin D
- Supplying daily requirement of vitamin D orally
  - For normal infant 400 IU/day
  - For preterm, twins 1000 IU/day

Thank you!