RICKETS IN CHILDREN

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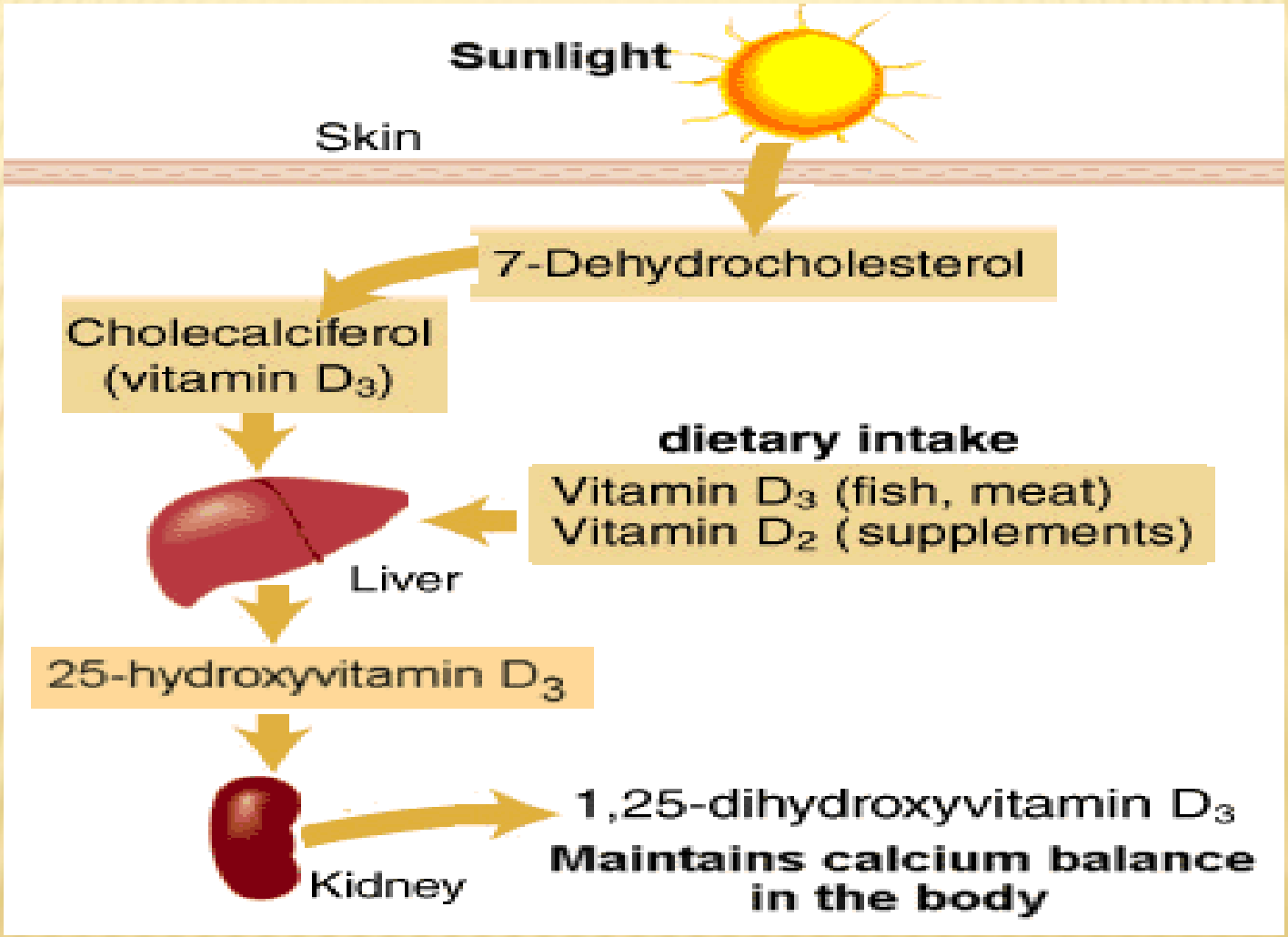
TITLE

- Vitamin D physiology
- Introduction
- Etiology
- Clinical feature
- Radiology
- Diagnosis
- Lab
- Treatment

VITAMIN D PHYSIOLOGY



- Source: -Fish, liver and oil,
  - Human milk (30-40 IU/L)
  - Exposure to sun light
- Vitamin D requirement:

  Infants- 200IU/day (5mcg)

  Children- 400IU/day (10mcg)

INTRODUCTION

Disease of growing bone due to unmineralized matrix at the growth plates and occurs in children only before fusion of epiphyses

ETIOLOGY

- VITAMIN D DISORDERS
- Nutritional vitamin D deficiency
  - Congenital vitamin D deficiency
  - Secondary vitamin D deficiency Malabsorption
  - Increased degradation
  - Decreased liver 25-hydroxylase

  -Vitamin D–dependent rickets type 1
  -Vitamin D–dependent rickets type 2
  - Chronic renal failure

CALCIUM DEFICIENCY

- Low intake
- Diet

  Premature infants (rickets of prematurity)

- Malabsorption
- Primary disease

  - Dietary inhibitors of calcium absorption

PHOSPHORUS DEFICIENCY

- Inadequate intake
- Premature infants (rickets of prematurity)
- Aluminum-containing antacids

RENAL LOSSES

- X-linked hypophosphatemic rickets
- Autosomal dominant hypophosphatemic rickets
- Autosomal recessive hypophosphatemic rickets
- Hereditary hypophosphatemic rickets with hypercalciuria
- Overproduction of phosphatonin
- Tumor-induced rickets
- McCune-Albright syndrome
- Epidermal nevus syndrome
- Neurofibromatosis
- Fanconi syndrome
- Dent disease

CLINICAL FEATURES OF RICKETS

- General
  - Failure to thrive
  - Listlessness
  - Protruding abdomen
  - Muscle weakness (especially proximal)
  - Fractures

HEAD

- Craniotabes
- Frontal bossing
- Delayed fontanel closure
- Delayed dentition; caries
- Craniosynostosis

- CHEST
  - Rachitic rosary
  - Harrison groove
  - Respiratory infections and atelectasis

BACK

- Scoliosis
- Kyphosis
- Lordosis

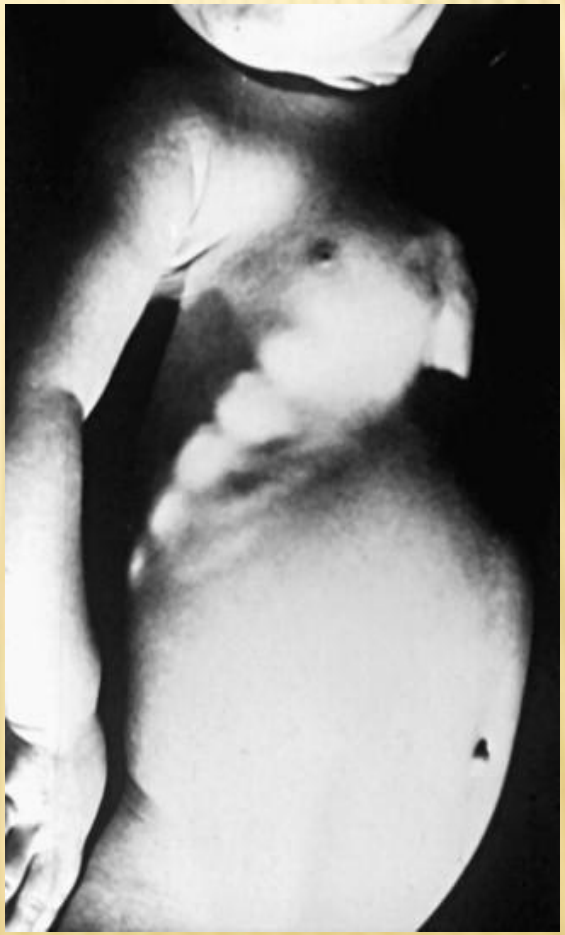
EXTREMITIES:

- Enlargement of wrists and ankles
-Valgus or varus deformities
-Windswept deformity (combination of valgus deformity of 1leg with varus deformity of the other leg)
-Anterior bowing of the tibia and femur
-Coxa vara
-Leg pain

HYPOCALCEMIC SYMPTOMS

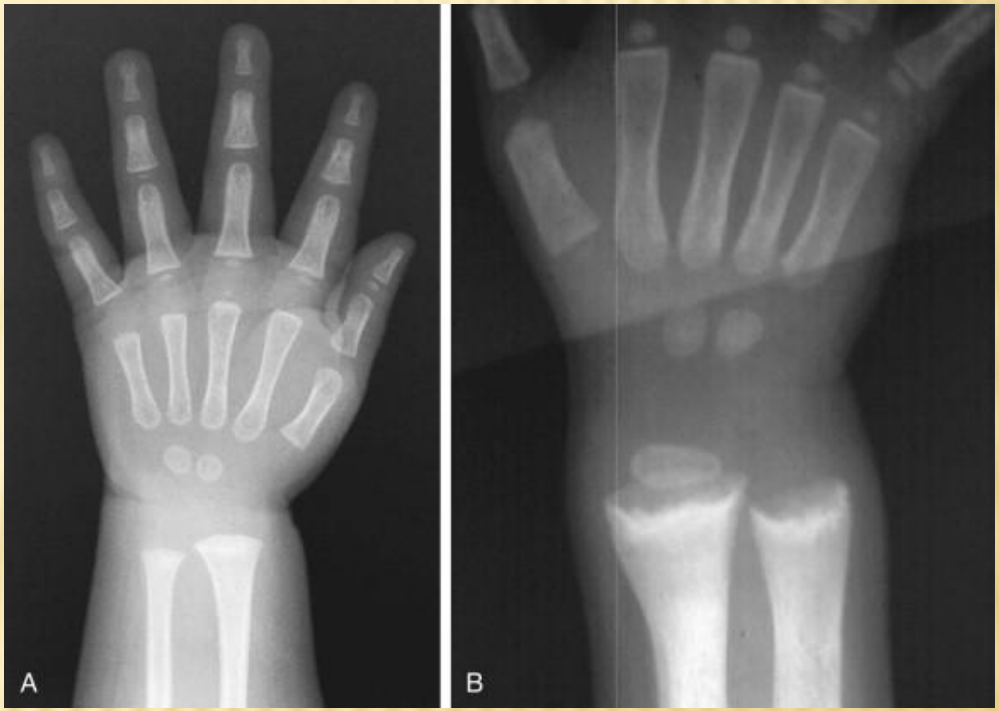
- Tetany
- Seizures
- Stridor due to laryngeal spasm

Deformities showing curvature of the limbs, potbelly, and Harrison groove.



RADIOLOGY

Wrist x-rays in a normal child (A) and a child with rickets (B). Child with rickets has metaphyseal fraying and cupping of the distal radius and ulna.



CLINICAL EVALUATION

- Dietary history
- Cutaneous synthesis
- Maternal risk
- Medication
- Malabsorption
- Renal disease
- Family history
- Physical Examination
- Lab Test

NUTRITIONAL VITAMIN D DEFICIENCY

- Vitamin D deficiency is most common cause of rickets globally
- Most common in infancy
- Transplacental transport of vit D provide enough vit D for first 1 to 2 months of life.
- Skin pigmentation

LABORATORY FINDINGS

Elevated:

- Alkaline phosphatase
- Parathyroid hormone
- Dihydroxyvitamin D

Decreased:

- Calcium
- Phosphorus
- Hydroxyvitamin D

Disorder	Ca	Pi	PTH	25-(OH)D	1,25-$(OH)_2$D	ALK PHOS	URINE Ca	URINE Pi
Vitamin D deficiency	N, ↓	↓	↑	↓	↓, N, ↑	↑	↓	↑
VDDR, type 1	N, ↓	↓	↑	N	↓	↑	↓	↑
VDDR, type 2	N, ↓	↓	↑	N	↑↑	↑	↓	↑
Chronic renal failure	N, ↓	↑	↑	N	↓	↑	N, ↓	↓
Dietary Pi deficiency	N	↓	N, ↓	N	↑	↑	↑	↓
XLH	N	↓	N	N	RD	↑	↓	↑
ADHR	N	↓	N	N	RD	↑	↓	↑
HHRH	N	↓	N, ↓	N	RD	↑	↑	↑
ARHR	N	↓	N	N	RD	↑	↓	↑
Tumor-induced rickets	N	↓	N	N	RD	↑	↓	↑
Fanconi syndrome	N	↓	N	N	RD or ↑	↑	↓ or ↑	↑
Dietary Ca deficiency	N, ↓	↓	↑	N	↑	↑	↓	↑

TREATMENT

- Stoss therapy – 300000 – 600000 IU Vitamin D oral or IM, 2-4 doses over one day
- Alternatively high dose vit D, 2000-5000 IU/day over 4-6 wk
- Followed by oral Vit D :

  < 1 year of age - 400IU

  > 1 years of age- 600IU
- Symptomatic hypocalcemia – IV calcium gluconate 100 mg/kg followed by oral calcium or calcitrol - 0.05mcg/kg/day

PROGNOSIS

- Most of children have excellent prognosis
- Severe disease causing permanent deformity and short stature

PREVENTION

- Daily multivitamin contain- 400IU vit D for infants while 600 IU/day for older children

SECONDARY VITAMIN D DEFICIENCY

- GI diseases - Cholestatic liver disease,
  - Cystic fibrosis, pancreatic dysfunction,
  - Defects in bile acid metabolism,
  - Celiac disease, Crohn disease. intestinal
  - lymphangiectasia
  - Intestinal resection.
- Severe liver disease decreases 25-D formation due to insufficient enzyme activity
- vitamin D deficiency due to liver disease usually requires a loss of >90% of liver function.
- Medication- Phenobarbital or phenytoin
  - isoniazid or rifampin.

TREATMENT

- high doses of vitamin D- 25-D
  (25-50 g/day or 5-7g/kg/day)
- 1,25-D, or with parenteral vitamin D.
- Degradation of vitamin D by the CYP system
  - Acute therapy as for nutritional deficiency
  followed by
- long-term administration of high doses of vitamin D
  - 1,000 IU/day) as much as 4,000 IU/day

VITAMIN D–DEPENDENT RICKETS, TYPE 1

- Autosomal recessive disorder
- Mutations in the gene encoding renal 1α-hydroxylase
- 1st 2 yr of life
- Classic features symptomatic hypocalcemia
- Normal levels of 25-D
- Low or normal levels of 1,25-D
- Renal tubular dysfunction- Metabolic acidosis and generalized aminoaciduria
- Teatment- 1,25-D (calcitriol)- 0.25-2 g/day

VITAMIN D–DEPENDENT RICKETS, TYPE 2

- Autosomal recessive disorder
- Mutations in gene encoding vitamin D receptor
- Levels of 1,25-D are extremely elevated

- Present during infancy, might not be diagnosed until adulthood.
- 50-70% of children have **alopecia,** range from alopecia areata to alopecia totalis.
- Epidermal cysts are less common

THANK YOU