RICKETS IN CHILDREN

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DNB Pediatrics
Vitamin D physiology
Introduction
Etiology
Clinical feature
Radiology
Diagnosis
Lab
Treatment
VITAMIN D PHYSIOLOGY

Sunlight
Skin

7-Dehydrocholesterol
Cholecalciferol (vitamin D₃)

Liver

25-hydroxyvitamin D₃

Kidney

1,25-dihydroxyvitamin D₃

Maintains calcium balance in the body

dietary intake
Vitamin D₃ (fish, meat)
Vitamin D₂ (supplements)
- 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
- 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
- Scoliosis
- Kyphosis
- Lordosis

EXTREMITIES:
- Enlargement of wrists and ankles
- Valgus or varus deformities
- Windswept deformity (combination of valgus deformity of 1 leg 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.
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
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
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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
- Treatment- 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