Board Review - Vitamins!
An 8-month-old infant presents with the primary complaint of irritability. He has been exclusively breastfed since birth. His mother was not interested in providing any supplemental foods because her milk supply has been adequate. Physical examination reveals a fussy infant who has frontal bossing and whose weight and height are both at the 25<sup>th</sup> percentile. The infant becomes irritable with movement of the left arm. Arm radiography reveals a humeral fracture and bowing of both radii. Chest radiography demonstrates enlargement of the costochondral joints.

Of the following, the MOST likely diagnosis is

A. Congenital syphilis
B. Osteogenesis imperfecta
C. **Vitamin D-deficient rickets**
D. Vitamin D-resistant rickets
E. Vitamin E deficiency
Know the absorption, storage, and metabolism of fat soluble vitamins (vitamin D)

Vitamin D metabolism

Metabolic activation of vitamin D to calcitriol and its effects on calcium and phosphate homeostasis. The result is an increase in the serum calcium and phosphate concentrations.
A 9-month-old exclusively breastfed baby presents with a seizure. A chest radiograph obtained to rule out aspiration pneumonia reveals rachitic changes of the ribs.

Of the following, the MOST likely serum laboratory findings are

A. Low calcium and elevated phosphorus
B. Low calcium and low phosphorus
C. Low calcium and normal phosphorus
D. Normal calcium and elevated phosphorus
E. Normal calcium and low phosphorus
Know the laboratory and radiologic findings in vitamin-D deficiency rickets

› Labs:
  – Low 25(OH)-vit D
    › “Deficient” = $<37.5 \text{ nmol/L}$
    › “Severely deficient” = $<12.5 \text{ nmol/L}$
  – Low calcium
  – Low phosphate
  – Elevated alkaline phosphatase
  – Elevated PTH

› Radiologic findings:
  – Frontal bossing
  – Widened cranial sutures
  – Craniotabes
  – Bowing of lower extremities
    › Widening of metaphyseal region
  – Kyphosis
  – Pelvic abnormalities
  – Osteopenia/Osteomalacia
<table>
<thead>
<tr>
<th>Type</th>
<th>Etiology</th>
<th>Calcium</th>
<th>Phosphorus</th>
<th>Parathyroid Hormone</th>
<th>1,25(OH)-D</th>
<th>Alkaline Phosphatase</th>
</tr>
</thead>
<tbody>
<tr>
<td>Vitamin D deficiency</td>
<td>Inadequate intake/little UV exposure</td>
<td>↓</td>
<td>↓</td>
<td>↑</td>
<td>Normal/↓</td>
<td>↑</td>
</tr>
<tr>
<td>Vitamin D–dependent (type I)</td>
<td>Low 1–α hydroxylase</td>
<td>↓</td>
<td>↓</td>
<td></td>
<td>↓</td>
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<tr>
<td>Vitamin D–dependent (type II)</td>
<td>1,25(OH)-D resistance</td>
<td>↓</td>
<td>↓</td>
<td>↑</td>
<td>↑</td>
<td>↑</td>
</tr>
<tr>
<td>Vitamin D–resistant (X-linked hypophosphatemia)</td>
<td>Defect of renal tubular phosphate reabsorption</td>
<td>Normal</td>
<td>↓</td>
<td>Normal</td>
<td>Normal/↓</td>
<td>↑</td>
</tr>
<tr>
<td>Renal disease</td>
<td>Defect of renal phosphate excretion</td>
<td>Mild</td>
<td>↓</td>
<td>↑</td>
<td>↑</td>
<td>↑</td>
</tr>
</tbody>
</table>

 Symptoms:

- Asymptomatic
- Failure to Thrive
- Hypotonia
- Seizures
- Tetany (hypocalcemia)
- Developmental delay
- Delayed tooth eruption

Recognize the presenting signs and symptoms of rickets...

Recognize the effects of vitamin-D deficiency in children of various ages
Prevention of vitamin D deficiency:

- Dietary sources - meat and fish, fortified foods
- Infant formulas have 400 IU/L
  > If formula fed and do not consume >1L of formula per day, supplement with 400 IU/day of vitamin D beginning a few days after birth.
    - *Darker skinned babies or those at high altitudes may require 800 IU/day.
    - *Premature infants may require up to 800 IU/day
- Sun exposure (no safe amount defined)
Recognize the presenting signs and symptoms of rickets, and manage appropriately ~&~ Plan the treatment of a patient with rickets

Treatment for deficiency/rickets:
  - Begin treating for deficiency at 25(OH)-vit D level of 37.5 nmol/mL-50 nmol/mL
    › Infants <1 month old: 1,000 IU/day
    › Infants 1-12 months old: 1,000-5,000 IU/day
    › Children >12 months old: 5,000-10,000 IU/day until radiologic evidence of healing (should be 2-4 weeks)
  - Maintenance therapy:
    › 400-1,000 IU/day
  - Non-compliance:
    › Can do 100,000-600,00 IU over 1-5 days
  - Concurrent calcium supplementation!

Understand the necessity of adequate vitamin D intake in children and adolescents
Hypervitaminosis D

› Recognize the signs and symptoms of hypervitaminosis D

  Hypotonia
  Anorexia
  Polydipsia
  Dehydration
  Hypertension
  Corneal clouding

› 25 (OH)-D level > 375 nmol/L
› Occurs when receiving >50,000 IU/Day
Vitamin D- Just the Facts!

› Know that breast milk is deficient in vitamin D

› Know that rickets may develop in rapidly growing premature infants with low intake of either calcium or phosphorous

› Know that a breast-fed infant with dark skin is susceptible to rickets
A 5-month-old male infant presents to your office because of pallor and irritability. He was born at term following an uncomplicated pregnancy and delivery and had a birth weight of 3150 grams. Because of newborn screening results, a sweat chloride examination was performed at 1 month of age that confirmed the diagnosis of cystic fibrosis. Since birth, the baby has been exclusively breastfed. At the time of diagnosis, pancreatic enzyme supplementation was begun and now includes 8,000 units of lipase per nursing session. Approximately 1 week ago, the mother noted that the baby was “breathing fast” and appeared very pale. On physical examination today, the well-developed infant has a weight of 6 kg, heart rate of 160 beats/min, and respiratory rate of 40 breaths/min. You also note conjunctival and mucous membrane pallor and a liver edge palpable 2 cm below the right costal margin. Laboratory results include:

- Hemoglobin 9 g/dL, White blood cell count 9.0x10^3/mcL, Albumin 3.8 g/dL, Reticulocyte count 12.5%

A blood smear demonstrates polychromasia and numerous schitocytes.

Of the following, the MOST appropriate treatment for this infant is

A. Alpha-tocopherol  
B. Ascorbic acid  
C. Cyanocobalamin  
D. Folic acid  
E. Thiamine

Fat Soluble- Vitamin A, D, E, K
Question 10

Which statement about Vitamin E is FALSE?

A. Vitamin E deficiency leads to loss of vibratory and position sense
B. Green leafy vegetables, vegetable oils, milk, and eggs are a good source of vitamin E
C. Risk factors for vitamin E deficiency include fat malabsorption and cholestatic liver disease
D. **Vitamin E is absorbed in the duodenum**
E. Vitamin E excess will present with nausea and diarrhea
 › Know the absorption, storage, and metabolism of fat soluble vitamins (vitamin E)

- Green, leafy veggies
- Vegetable oils
- Milk
- Eggs

Pre-term infants have decreased amounts of intraluminal bile acids → decreased absorption of triglycerides/fat-soluble vitamins → different abilities in preterm infants to digest fat/absorb vits
Know the signs, symptoms, and causes of vitamin E deficiency

› **Signs/Symptoms:**
  - Myopathy
  - Ataxia
  - Pigmented retinopathy with vision loss
  - Loss of vibratory/position sense
  - Hyporeflexia
  - Generalized weakness
  - Hemolytic anemia

› **Causes:**
  - Common or Uncommon?
  - Fat malabsorption
  - Short bowel syndrome
  - Cholestatic liver disease
  - Prematurity
Vitamin E deficiency

- Deficiency < 5 mg/L
  - Nausea, diarrhea, vitamin K antagonism

- Treated with:
  - 15-25 mg/kg/day vitamin E supplementation
A term infant who was delivered at home is breast-feeding poorly. She is referred to you 7 days after birth by a lactation consultant because of bleeding from the umbilical cord and blood in the stool. Findings on the physical examination are normal. There are no petechiae, and there is no hepatosplenomegaly, purpura, or neurologic abnormality.

Of the following, the MOST likely diagnosis is

A. Classic vitamin K deficiency
B. Disseminated intravascular coagulation
C. Factor VIII deficiency
D. Neonatal alloimmune thrombocytopenia
E. Von Willebrand disease
Know the absorption, storage, and metabolism of fat-soluble vitamins (K)

Cofactor for gamma-glutamyl carboxylase

Coagulation factors:
Factor II, VII, IX, X, protein C, protein S

Is it transported across the placenta?
Recognize the clinical manifestations of vitamin K deficiency

› Vitamin-K deficiency bleeding (VKDB)
  – Previously hemorrhagic disease of the newborn
    › Early
    › Classic
    › Late
  – Classic labs?
    › INR > 4
    › Elevated PT
    › Normal bleeding time
    › Normal platelets
    › Normal fibrinogen
    › Correct with vitamin K admin
Vitamin K deficiency bleeding

› Early
  – 1st 24 hours of life
  – Presentation: Cephalohematomas, intra-abdominal hemorrhage, intracranial bleeding
  – Usually in babies born to mothers on anti-vitamin K drugs (carbamazepine, phenytoin, barbiturates, isoniazid, rifampin, cephalosporins, warfarin)

› Classic
  – Day 2-7 of life
  – Presentation: Umbilical bleeding, GI tract bleeding, oozing from puncture sites, intracranial bleeding
  – Delayed vitamin K administration, lack of dietary vitamin K
Vitamin K deficiency bleeding

› Late
  – 2-12 weeks of life
  – Presentation: Significant intracranial bleeding (50% of patients)
  – Exclusively breastfed infants without vitamin K supplementation

› Treatment:
  – Prevent it! → all newborns to receive Vitamin K 1 mg IM
  › Can do oral supplementation but not proven to be effective
Total Specs Covered: 14+

Happy Holidays Everyone!!! 😊