Nutritional Deficiencies

Kristina Burke, MD, and Erin Adams, MD

Nutritional Deficiency	Setting	Findings	Miscellaneous
Fat Soluble Vitamins			
Vitamin A Deficiency **Phrynoderma**	Diseases/states of fat malabsorbtion (Crohn's, celiac, CF, cholestatic liver dz, bypass surgery, pancreatic insufficiency)	- Skin: Toad skin, keratotic papules over extremities, back, abdomen and buttocks. - Eyes: nyctalopia, keratomalacia, xerosis corneae, xerophthalmia, white conjunctival spots (Bitot spots)	- Increased mortality from inflammatory disease of gut and lungs - Children with measles (rubeloa) require Vit A supplementation - Visual symptoms resolve quickly with supplementation, corneal scarring is permanent
Vitamin A excess	Kids at higher risk; megavitamin supplementation; patients with liver disease or on dialysis	Hair loss, exfoliative cheilitis, generalized exfoliation, loss of eyebrows, bone growth retardation, pseudotumor cerebri	- Fatigue, myalgia, depression - Stop all Vit A supplementation if on synthetic retinoid
Vitamin D deficiency	Diseases/states of fat malabsorbtion; Anticonvulsants; elderly; patients with photosensitive disorders	Alopecia	- Kids – rickets - Adults – osteomalacia - Schimmelpenning syndrome – Vit D-resistant -rickets
Vitamin D excess	Continued supplementation	No skin findings	- Hypercalcemia/calciuria, anorexia, vomiting, diarrhea
Vitamin E		Peripheral edema, neuromyopathy	
Vitamin K deficiency	Diseases/states of fat malabsorption; biliary disease, cholestasis of pregnancy or liver disease; Drugs: coumarin, cephalosporins, cholestyramine, salicylates	Purpura, hemorrhage, ecchymosis. Decreased factors II, VII, IX, and X	- Coumarin, cephalosporins, salicylates inhibit Vit K epoxide reductase - In adults, usually synthesized in adequate amounts by gut flora
Water Soluble Vitamins			
Vitamin C deficiency **Scurvy**	Alcoholics Restricted diets Psychiatric patients	Four H's: Hemorrhagic signs, hyperkeratosis of hair follicles, hypochondriasis, hematological abnormalities. Specifically, perifollicular petechiae, hemorrhagic gingivitis, epistaxis, corkscrew hairs	- Symptoms develop ~3mo after deficiency begins - Pseudoparalysis (due to subperiosteal hemorrhage) - Delayed wound healing
Vitamin B1 deficiency (Thiamine) **Beriberi**	Alcoholics Polished-rice diets Pregnancy (esp hyperemesis gravidarum)	Dry beriberi: edema and red, burning tongue, nervous system defect (peripheral neuropathy, Wernicke-Korsakoff syndrome),	Wet beriberi – high output cardiac failure
Vitamin B2 deficiency (Riboflavin)	Alcoholics, acute boric acid ingestion, hypothyroidism, neonatal phototherapy, chlorpromazine	Oro-oculo-genital syndrome: seb derm-like changes and fissuring of perioroficial and genital areas, perleche, cheilosis, depapillated glossitis (magenta tongue), conjunctivitis	- Dramatic response to riboflavin supplementation
Vitamin B3 deficiency (Niacin) **Pellagra**	Alcoholics, corn-rich diet, carcinoid tumors, Hartnup disease, GI disease (Crohn's), psych (anorexia) Meds: isoniazid, 5-FU, azathioprine, ethionamide, protionamide, pyrazinamide	3 D's: dermatitis, diarrhea, dementia. Dermatitis: photosensitive over dorsal hands, arms, face, neck (Casal's necklace); sulfur flakes over nose, copper hue to affected skin, hyperpigmentation	- Abdominal pain, weakness, diarrhea, depression - Can also result from tryptophan deficiency (precursor to niacin) - Rapid improvement (~24hrs) of symptoms once supplementation started
Vitamin B6 deficiency (Pyridoxine)	Uremia, cirrhosis Drugs: isoniazid, penicillamine, hydralazine, cycloserine	Seborrheic dermatitis-like, atrophic glossitis, angular cheilitis conjunctivitis, intertrigo. Occasionally pellagra-like.	- Somnolence, confusion, neuropathy.
Vitamin B12 deficiency (cyanocobalamin)	Malabsorption: decreased gastric intrinsic factor (pernicious anemia), gastrectomy, distal ileum resection, achlorhydria Drugs: metformin, antacids	Hyperpigmentation esp in flexures, palms, soles, nails. Tongue smooth red, atrophic and painful	- Weakness, parasthesias, ataxia - Liver has large body stores, deficiency develops 3-6 years after GI abnormalities - Megaloblastic anemia
Folic Acid (Vit B9) deficiency		Hyperpigmentation, glossitis, cheilitis	- Megaloblastic anemia - Neural tube defects
Biotin deficiency	**Genetic:** deficiency of holocarboxylase synthetase or biotinidase **Acquired:** short gut, malabsorbtion, avidin from egg whites	Alopecia, periorificial with patchy red, eroded lesions of face and groin, conjunctivitis, secondary infections (candida). Findings similar to zinc deficiency and essential fatty acid deficiency	- Limb parasthesias, weakness, depression, lethargy - Skin lesions resolve rapidly w/ supplementation, neurologic damage may be permanent
Zinc deficiency	**Genetic:** acrodermatitis enteropathica (defect in intestinal absorbtion of zinc – zinc transporter SLC39A4 mutation) **Acquired:** high-fiber foods (phytate), low maternal milk zinc levels, alcoholism, HIV	Pustular and bullous acral and periorificial dermatitis, angular chelitis, stomatitis, periungual scaling, nail dystrophy, alopecia, diarrhea	- Consider this in an infant with chronic diaper rash and diarrhea - Check Alk Phos (zinc dependent enzyme - can be low)
Copper deficiency	**Genetic:** Menkes kinky hair disease (XLR, mutations in ATP7A copper transporting APTase) **Acquired:** rare, malnutrition, zinc excess	Hair: pili torti, monilethrix, trichorrhexsis nodosa Diffuse pigmentary dilution Failure to thrive, lethargy	- Tyrosinase is copper dependent

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Nutritional Deficiencies (cont.)

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Water Soluble Vitamins			
Copper excess	**Genetic:** Wilson dz (AR, mutation in ATB7B copper transporter) **Acquired:** excessive intake	Kayser-Fleischer rings (copper deposition in Descemet's membrane), liver disease, neurological findings	- Penicillamine therapy may precipitate elastosis perforans serpiginosa
Iron deficiency	Hemorrhage, blood loss	Pallor of mucous membranes, koilonychia, glossitis (smooth, atrophic tongue), angular cheilitis, telogen effluvium	- Plummer-Vinson syndrome: microcytic anemia, dysphagia (esophageal web), glossitis in middle-aged women
Iron Excess	**Genetic:** Hemochromatosis (mutation in HFE gene C282Y and H63D) **Acquired**	Gray to brown mucocutaneous hyperpigmentation	- Tetrad: cirrhosis, diabetes, hyperpigmentation and heart failure - Avoid Vit C supplementation and raw seafood (V. vulnificans and Yersinia infections)
Selenium deficiency	Parenteral nutrition	Skin hypopigmentation and white nails (**pseudoalbinism**)	- Cardiomyopathy, muscle pain, weakness - Needed for glutathione peroxidase (protective against oxidative stress)
Essential Fatty acid deficiency	Low birth-wt infants, parenteral nutrition, inflammatory bowel disease, intestinal surgery	Generalized xerosis, widespread erythema, weeping intertrigenous eruption, hair lightens	- Eicosatrienoic acid-to-arachidonic acid ratio > 0.4 = diagnostic - EFA's constitute 13-30% of skin fatty acids
Calorie deficiency **Marasmus**	Protein-energy malnutrition Also, malignancies, HIV, restrictive diets	Skin: dry, wrinkled loose, hyperpigmentation and desquamation, excess of lanugo-like hair, "Monkey facies": loss of buccal fat pad, purpura	
Protein deficiency **Kwashiokor**	Rice-based diets, extensive GI surgeries, protein-loosing enteropathies	Hair: dry, hypopigmented, **"flag sign"** Skin: desquamation, hypopigmented in areas of friction, "flaky paint," "mosaic skin."	- Edema and potbelly - Mental status changes
Carotenemia	Too many carrots (squash, pumpkins, spinach, etc…) Hypothyroidism	Orange-yellow discoloration prominent in areas w/ abundant sebaceous glands (nasolabial folds/forehead), also palms & soles	
Lycopenemia	Too many tomatoes, beets, and chili beans	Reddish discoloration of skin	

Memory Tool for B-vitamins:
The – B1 (Thiamine)
Rich – B2 (Riboflavin)
Never – B3 (Niacin)
Pay – B6 (Pyridoxine)
Cash – B12 (Cyanocobalamin)

References:

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