MALABSORPTION SYNDROMES

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JUST

MISCONCEPTION

Chronic diarrhea = Malabsorption

EPIDEMIOLOGY AND PATHOPHYSIOLOGY

- Nutrient absorption is an efficient process that is regionalized in the gut and depends on Pancreatic ,Hepatobiliary secretory function and the mucosal integrity of the intestine
- Neonates and young infants have a reduced ability to digest fat *(healthy infants may have visible fat in their stools)*
- Coordinated intestinal peristalsis is important in facilitating digestion *(abnormalities are common in preterm infants and very ill Children)*

Digestive/Absorptive process categorized into:

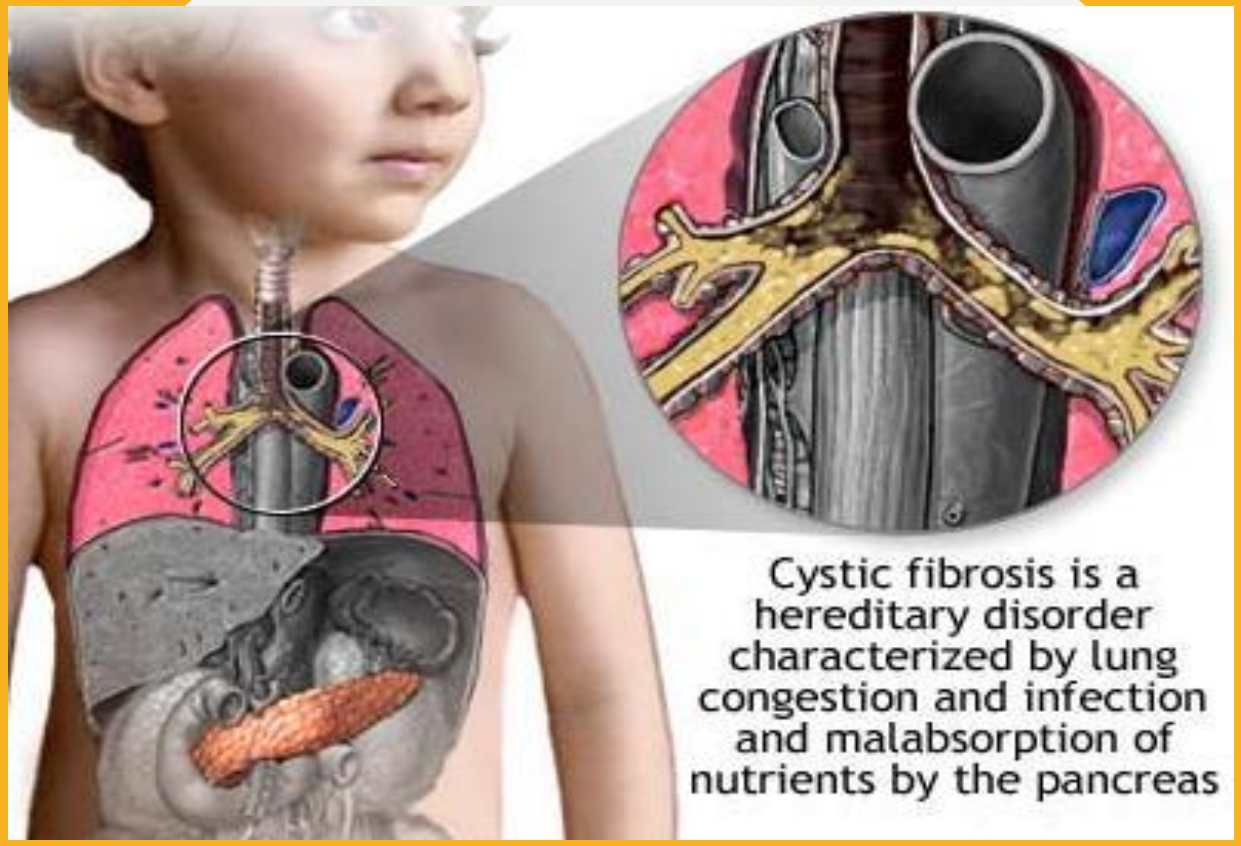
1. intraluminal
2. mucosal
3. venous and lymphatic transport phases

Most nutrient digestion and absorption occurs in the proximal small intestine

MALABSORPTION SYNDROMES

PANCREATIC DISORDERS

CYSTIC FIBROSIS



SHWACHMAN DIAMOND SYNDROME

A R Disorder

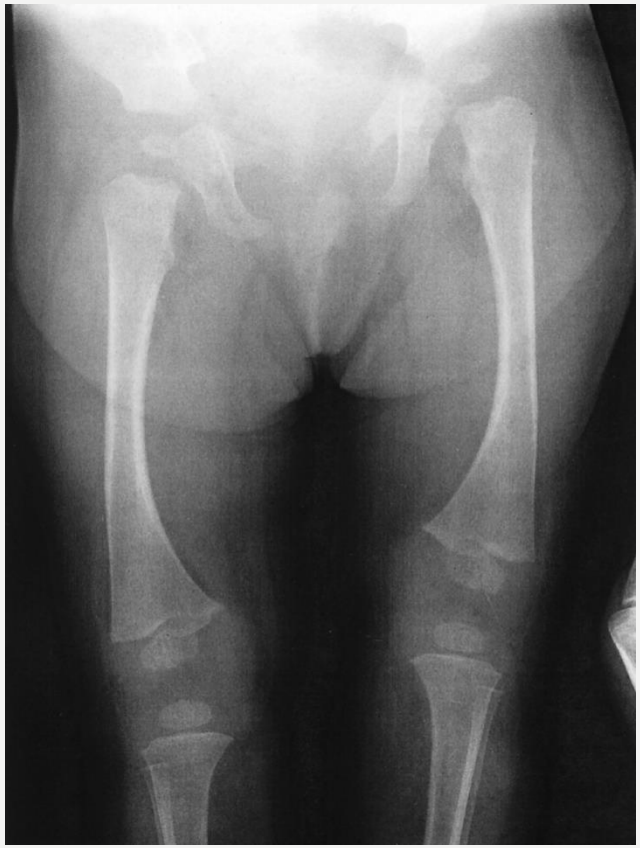
1 in 10,000 live births

Phenotypic features include:

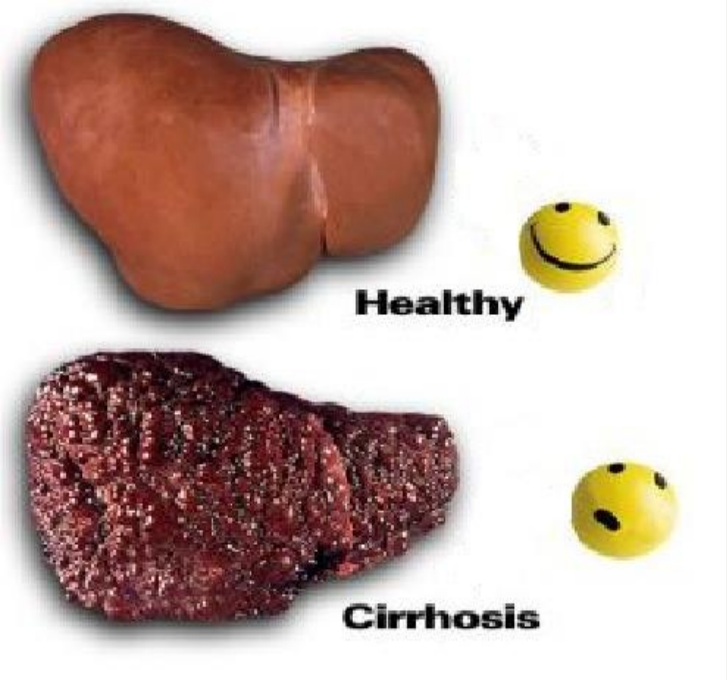
- short stature
- skeletal abnormalities
- recurrent infections
- various forms of bone marrow dysfunction: pancytopenia and cyclic neutropenia

- Pre-leukemic state

The pancreatic lipomatosis leading to insufficiency



HEPATIC DISORDERS



CHRONIC CHOLESTASIS

Chronic cholestasis is evident to the practitioner because of the child's enlarged liver and jaundice

Children with chronic cholestasis experience:

1. Growth failure
2. Chronic malnutrition
3. Fat soluble vitamin deficiencies

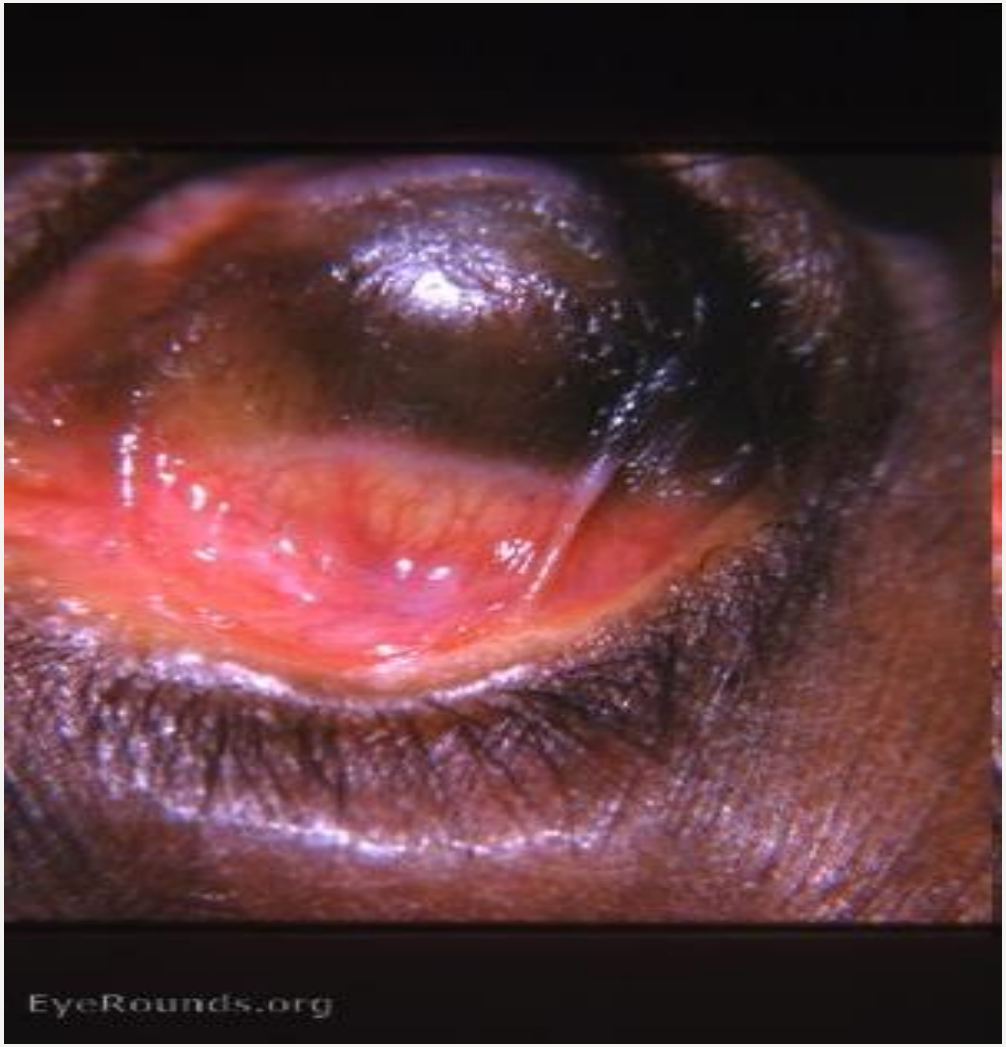
VITAMIN E DEFICIENCY

- Special concern because several interrelated digestive steps are necessary for absorption of this vitamin
- Signs of vitamin E deficiency include:

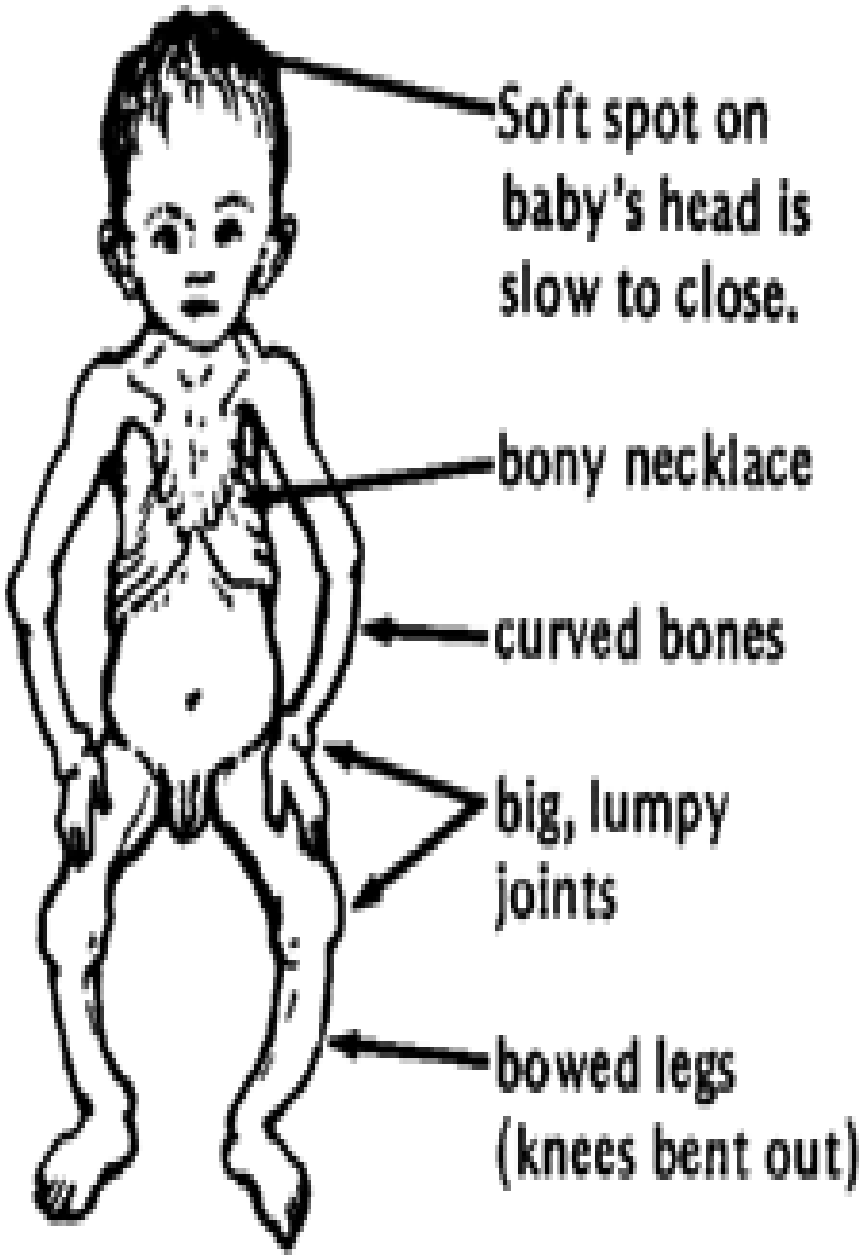
a. ataxia
b. decreased or absent deep tendon reflexes
c. ocular palsy
d. hemolytic anemia

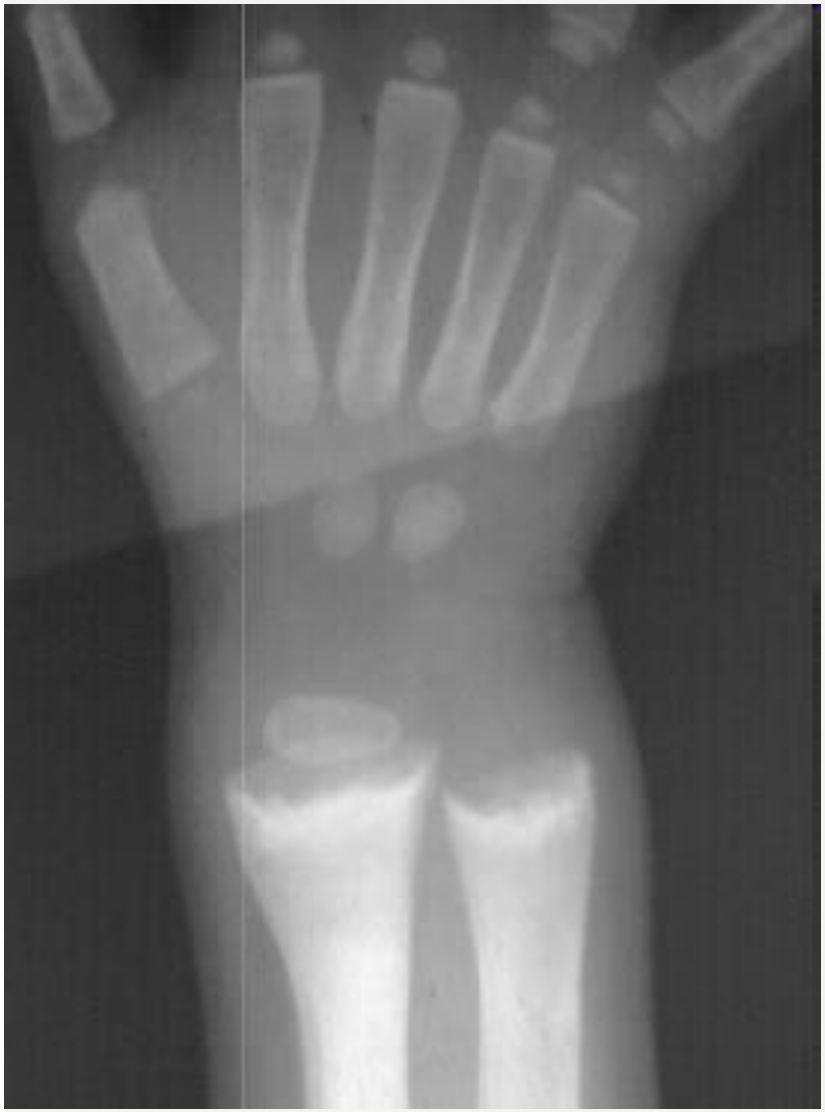
VITAMIN K MALABSORPTION

Lead to reduced hepatic synthesis of coagulation factors


EyeRounds.org

SIGNS OF RICKETS





Persistent cholestasis early infancy mostly related to biliary atresia

Rare conditions either syndromic or related to specific inherited bile duct epithelial transport defects could cause it

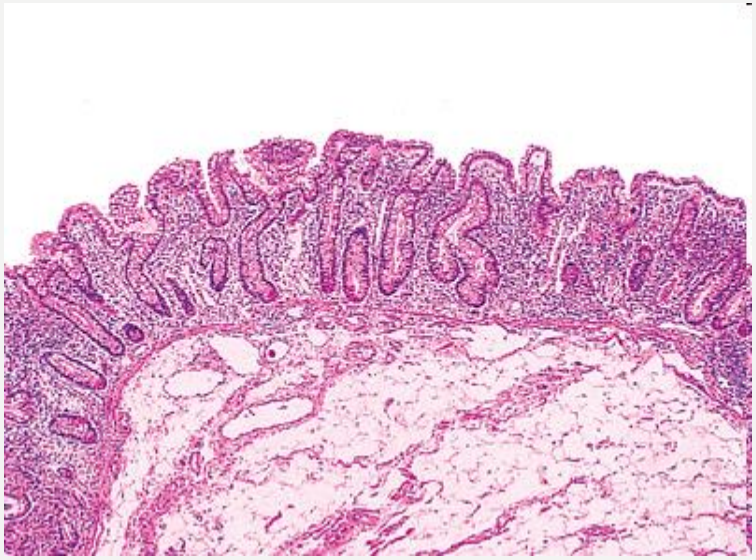
Broadened forehead, pointed chin, and elongated nose with bulbous tip

AD

Physically:

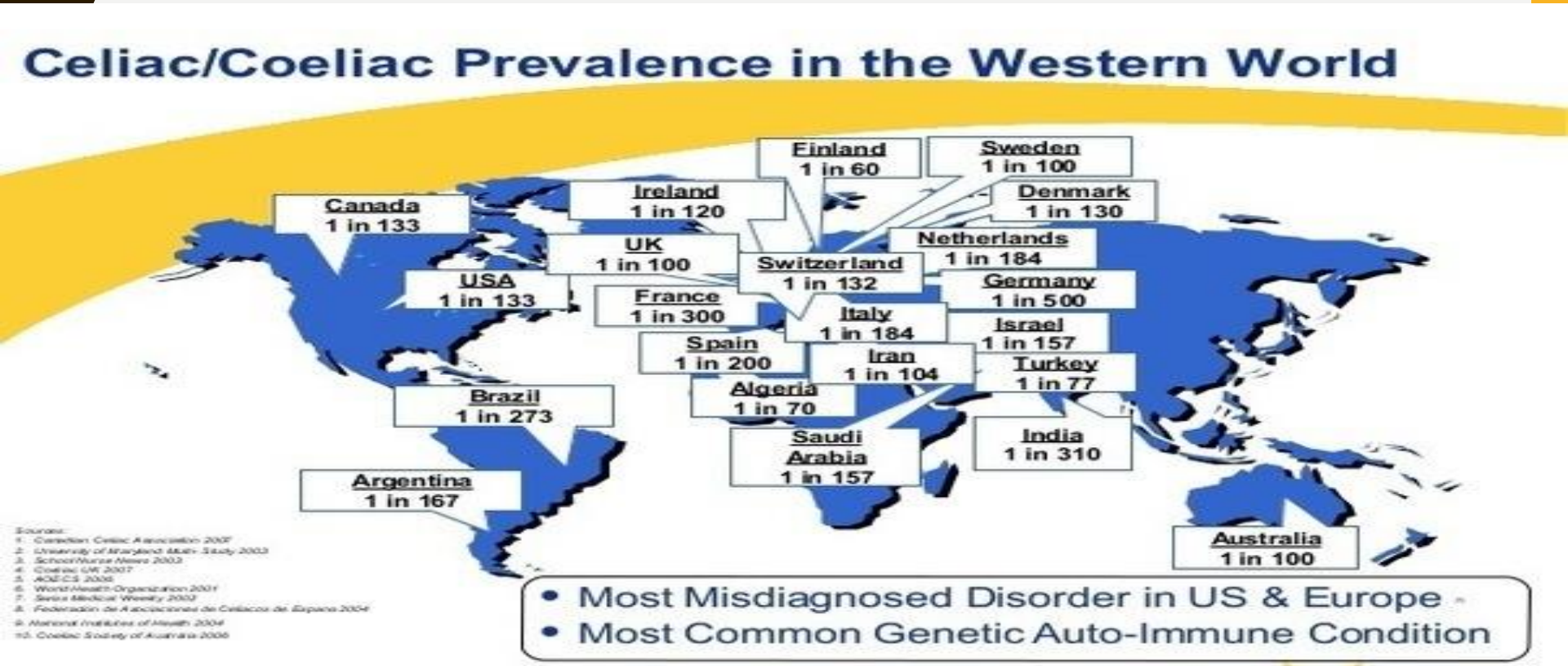
- Nutrition & Growth
- Head & Neck
- Ophthalmologic Ocular abnormalities
- Heart
- Hepatic
- Skeletal Abnormalities
- Neurological
- Renal
- Vascular abnormality

MUCOSAL SURFACE INJURY



CELIAC DISEASE

- Worldwide increasing incidence
- Frequency of 1 in 100 to 500
- Progressive inflammatory damage to small bowel mucosa, with the enteritis appearing to be most severe proximally



Appears in genetically susceptible individuals who sustain an autoimmune reaction to the toxic gliadin protein fraction of gluten

> 99% of celiacs are +ve for HLA-DQ2 or -DQ8

There is a strong trend for inheritance with certain identified *susceptibility risk factors*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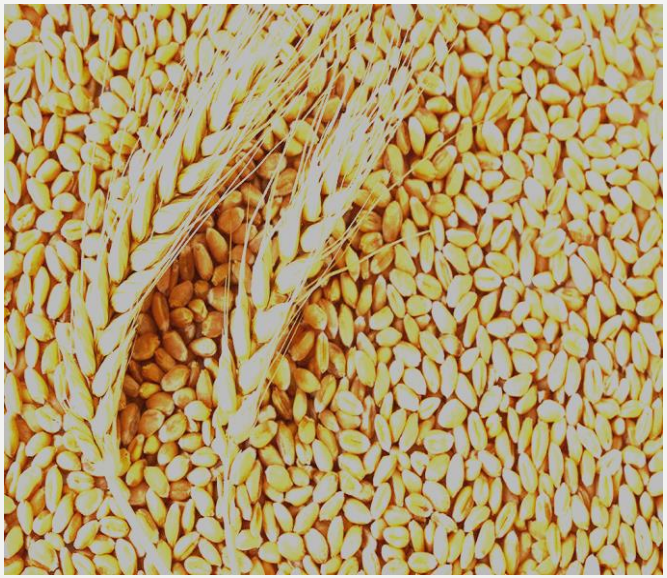
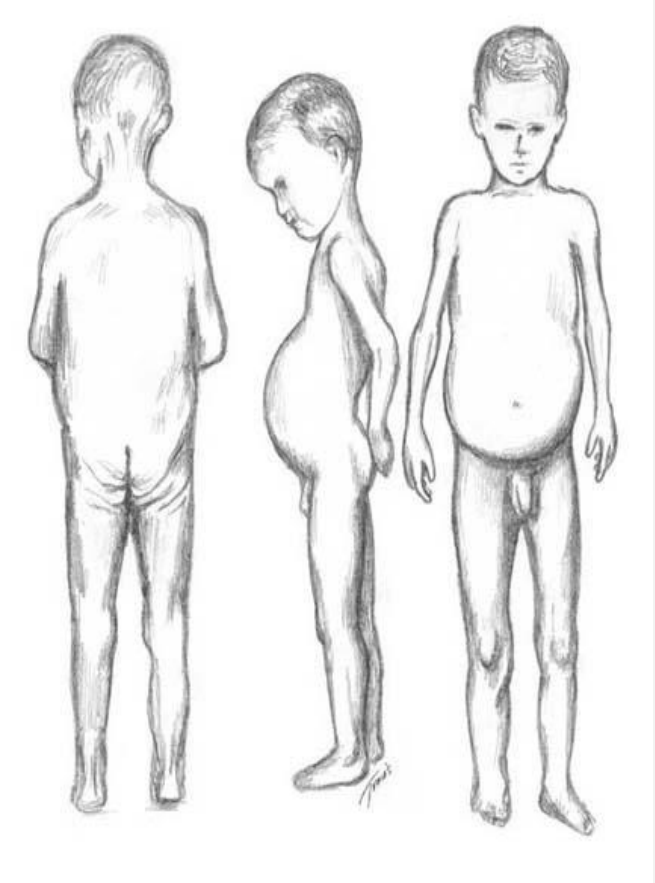


CLINICAL PRESENTATION

- The classic case is the infant who has a "potbelly," wasted extremities, bulky stools, irritability, and laboratory evidence of Malabsorption
- Diarrhea is not necessary, constipation may be evident
- There are more subtle presentations, asymptomatic forms, and delayed-onset disease

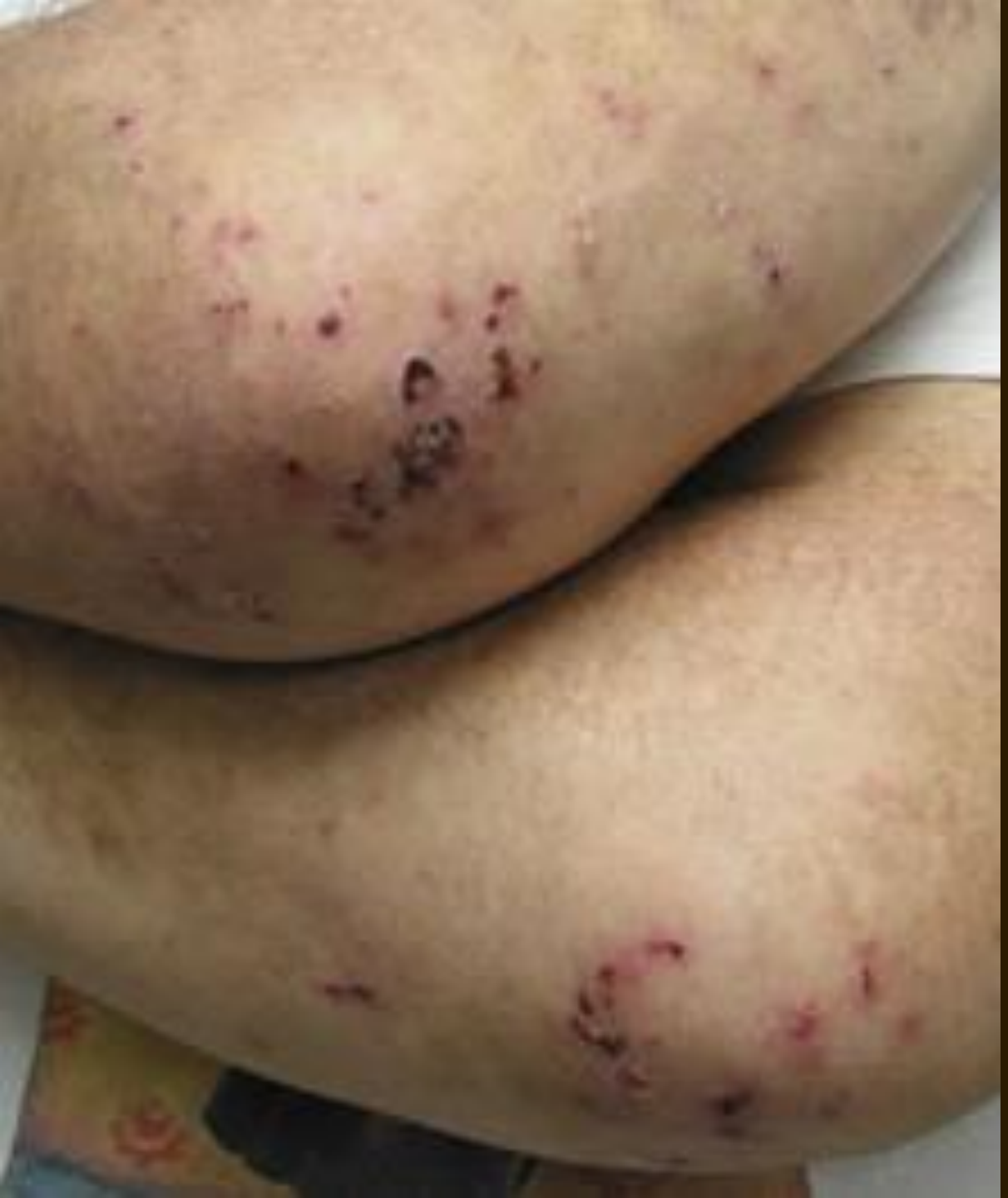


THE EXTRAINTESTINAL SYMPTOMS SECONDARY TO MALABSORPTION

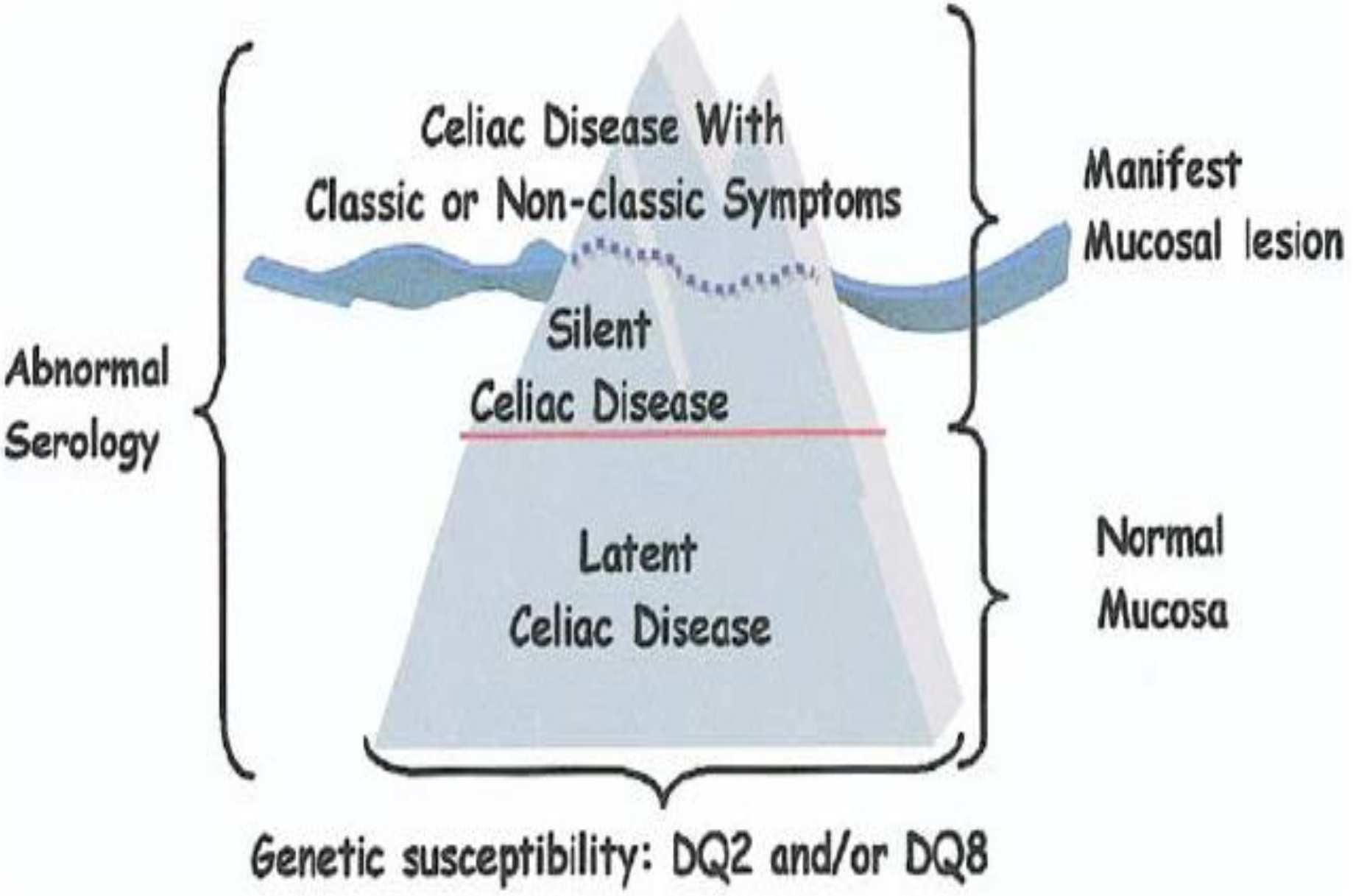
• Peripheral neuropathy	Vitamin B12 and B1 deficiency, Vit E
• Anemia	Iron, Vit. B12 and folate def.
• Growth failure in children	
• Bone pain	Osteoporosis and osteopenia, Vit. D and Ca def.
• Muscle cramps	Mg and Ca def.
• Night blindness	Vit. A def.
• Weight loss	Impaired absorption of most nutrients
• Edema	Protein and albumin loss
• Weakness	Hypokalemia and electrolyte depletion
• Bleeding and hematoma	Vitamin K deficiency

THE EXTRAINTESTINAL SYMPTOMS/MANIFESTATIONS NOT SECONDARY TO MALABSORPTION (ATYPICAL CD)

- *Neurological disorders* : depression, epilepsy, migraine, ataxia
- *Dermatitis herpetiformis*
- *Elevated liver enzymes, liver failure*
- *Infertility*
- *Stomatitis*
- *IgA nephritis*
- *Myocarditis*
- *Idiopathic pulmonary hemosiderosis*
- *Arthritis*



Dermatitis Herpetiformis

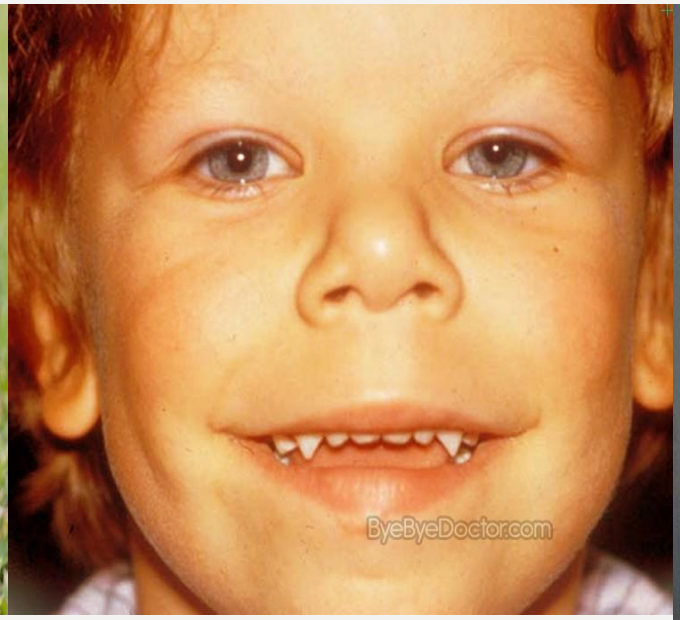

Celiac Disease With
Classic or Non-classic Symptoms
Silent
Celiac Disease
Latent
Celiac Disease
Abnormal
Serology
Manifest
Mucosal lesion
Normal
Mucosa
Genetic susceptibility: DQ2 and/or DQ8

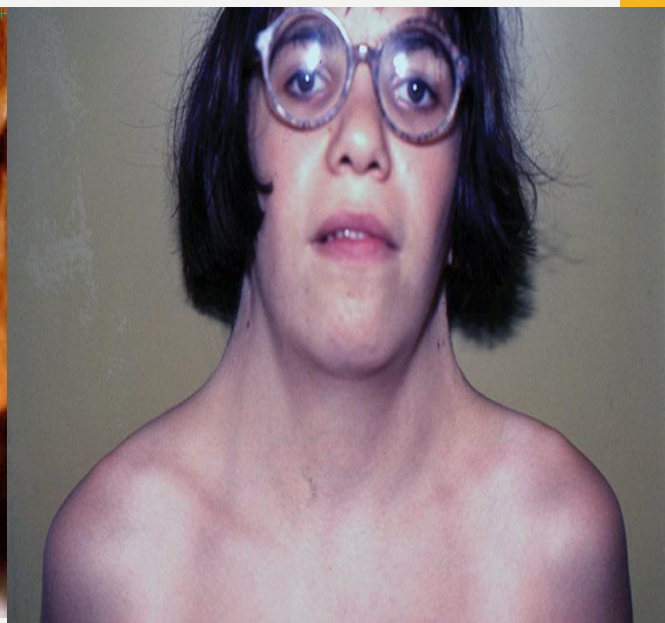
Clinical form	Histologic and clinical manifestations
CD with classic symptoms	Fully expressed enteropathy Intestinal symptoms
CD with nonclassic symptoms	Fully expressed enteropathy Extraintestinal manifestations
Silent	Fully expressed enteropathy Minimal complaints or symptom-free (occasionally discovered by serologic screening)
Potential	Minimal changes enteropathy or normal small intestinal mucosa Sometimes symptomatic

Increase incidence with other autoimmune disorders:

1. Type 1 diabetes
2. Autoimmune thyroid disorders
3. Rheumatoid Arthritis, and other vasculitic disorders
4. Autoimmune liver disease

Other Syndromes:





LABORATORY EVIDENCE

Complex anemia

Malabsorption of fat-soluble vitamins

Hypoproteinemia

Fecal α-1-antitrypsin

SCREENING BLOOD TESTS

Antiendomysial IgA , and anti-tissue transglutaminase (Transglutaminase 2) IgA *most sensitive and specific*

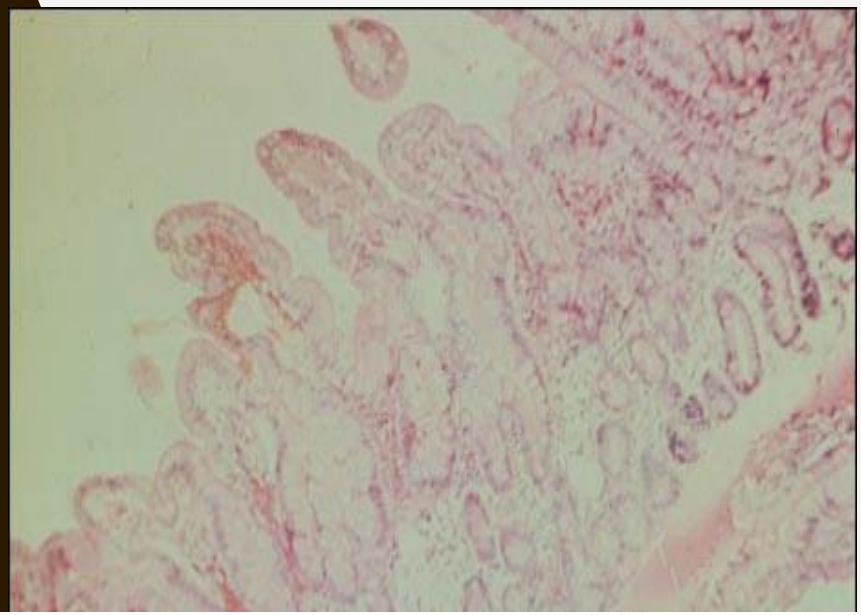
IgA deficiency ????!!!!!!!!!!

Anti-Gliadin Ab

DIAGNOSIS

- Signs and/or symptoms consistent with celiac disease
- Positive serology testing
- Characteristic changes of the duodenal mucosa
- Full and unequivocal clinical remission after withdrawal of gluten
- Disappearance of circulating antibodies

Small intestinal biopsy to confirm the presence of mucosal damage while the patient is on a gluten containing diet



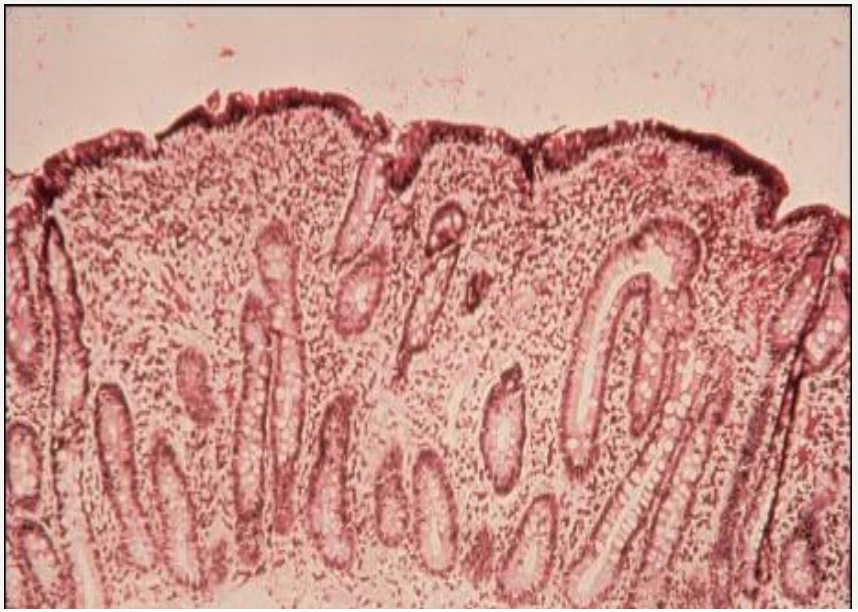


Table I. Histological classification for coeliac disease

	Marsh 0	*Marsh I*	*Marsh II*	*Marsh IIIa*	*Marsh IIIb*	*Marsh IIIc*
IEL/100 enterocytes	< 25/100EC	> 25/100 EC	> 25/100 EC	> 25/100 EC	> 25/100 EC	> 25/100 EC
Crypt hyperplasia	-	-	Hyper plastic	Hyper plastic	Hyper plastic	Hyper plastic
Villous atrophy	-	-	-	PVA	STVA	TVA
	Microscopic enteritis			Macroscopic enteritis		

EC: enterocytes; PVA: partial villous atrophy; STVA: subtotal villous atrophy; TVA: total villous atrophy.

Partial /non-compliance increased risk for gastrointestinal cancers and Extra intestinal medical problems

INITIAL EVALUATION OF SUSPECTED MALABSORPTION

1. **Detailed history and physical examination**
2. **Serially plotted points on growth chart**
3. **Complete blood count, ESR**
4. **Chemistry panel (albumin, total protein, liver enzymes, Ca, PO4 electrolytes, bilirubin)**
5. **Examination of stool and test for occult blood**

During the stool examination, it is common to see undigested food in the stools of
healthy infants and young children

SCREENING TESTS

Blood Tests
Inflammatory bowel disease panel **Immunoglobulin A level and celiac disease panel** **Cystic fibrosis DNA mutation** **Vitamin levels** **—Vitamin B12, folate,** **—Fat-soluble vitamin E,A, 25-OH D, prothrombin time)**

Stool Analysis

- FA1AT
- Stool trypsin
- Stain for dietary fat
- 3-day quantitative fecal fat
- pH and Clinitest for sugars

Other
•Sweat test •Hydrogen breath test for carbohydrate malabsorption

DEFINITIVE TESTS:

Gastrointestinal Endoscopy Biopsy

- **Special stains for celiac disease, infection (giardiasis, Cryptosporidium, mycobacteria)**
- **Enzymology**
- **Electron microscopy / ultrastructure**

Endoscopic Retrograde Cholangiopancreatography / Duodenal Intubation

Pancreatic enzyme analysis

Liver Biopsy

Cholestatic syndromes

Imaging Procedures

Liver scan

Radiolabeled Tc albumin lymphatic scan

Computed tomography scanning of liver and pancreas

Upper gastrointestinal radiographic series to rule out bowel stenosis or segmental dilation (bacterial overgrowth syndrome)

THANK YOU