INDEX

Biochemistry	05-26
Immunology	27-36
Microbiology	37-69
General Pathology	70-80
General Pharmacology	81-88
Biostatistics	89-97
Cardiology	98-120
Dermatology	121-132
Endocrine system	133-148
GIT	149-172
Hematology	173-193
Musculoskeletal system	194-225
Neurology	226-259
Psychiatry	260-279
Renal System	280-298
Reproductive System	299-322
Respiratory System	323-335
Behavioural Science	336-358

ABBREVIATIONS AND SYMBOLS

In risk factors, remember them in the order of preference

***	-	Very important in step 1 Examination.
AMS	-	Altered Mental Status
ABGA	-	Arterial Blood Gas Analysis
B/L	-	Bilateral
CF	-	Clinical features
C/O	-	Complications
CXR	-	Chest X Ray
DOC	-	Drug of Choice
Dx	-	Diagnosis
Etiology	-	Etiology
HRT	-	Hormone Replacement therapy
I & D	-	Incision and Drainage
M/E	-	Microscopic Examination
NBS	-	Next Best Step
OCP	-	Oral Contraceptives
PATH	-	Pathogenesis
Ppx	-	Prophylaxis
PPROM	-	Preterm Premature Rupture Of Membranes
Risk factor	-	Risk factors
ROM	-	Rupture Of Membranes
RUE	-	Right Upper Extremity
S /E	-	Side Effects
Rx	-	Treatment
U/L	-	Unilateral
UPT	-	Urine Pregnancy Test
USG	-	Ultrasonography
y/o or yr	-	year old

IMPORTANT NOTES

We advise you to remember the following important laboratory values to help manage time effectively during the lengthier blocks in the USMLE STEP 1 examination.

Sodium (Na): 135-145 mEq/L
Potassium (K): 3.5-5.0 mEq/L
Bicarbonate (HCO3): 22-28 mEq/L

Hematocrit (Hct): 40-50% (men), 35-45% (women)
Platelets: 150,000-450,000/mcL

Prothrombin Time (PT): 11-13.5 seconds
Activated Partial Thromboplastin Time (aPTT): 25-35 seconds
Bleeding time - 2-7 minutes

Aspartate Aminotransferase (AST): 10-40 U/L
Alanine Aminotransferase (ALT): 7-56 U/L

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BIOCHEMISTRY

1. Alcoholic patient with just peripheral neuropathy?

Vitamin B6 deficiency

Important functions :

- Used in heme synthesis.
- Used in GABA synthesis (hence B6 deficiency can lead to seizures).
- Used in synthesis of dopamine, serotonin, norepinephrine, epinephrine.

Most important reasons for B6 deficiency : Malnutrition, heavy Alcohol use, Isoniazide.

Differential :

a) Vitamin B1 deficiency (wernicke korsakoff) = also look for other symptoms like confusion, ataxia, memory changes, visual disturbances, etc).

b) Vitamin B12 deficiency = look for other symptoms like loss of vibration, proprioception, ataxia, etc (Subacute combined degeneration).

2. Recurrent infections + other possible symptoms are failure to thrive (body weight <= 5th percentile), lung infection, bronchiectasis, steatorrhea, fat soluble vitamin deficiency (A,D,E,K) ?

Cystic fibrosis

Which channels are dysfunctional ? **- ATP gated Chloride channels**

Which gene is mutated ? - CFTR (most commonly = delta F508 → leading to deficiency of Phenylalanine).

Inheritance : Autosomal recessive

Pathogenesis : Gene mutation → absent chloride channel on plasma membrane.

In sweat glands : There is inability to reabsorb Cl- from lumen of the gland → Increased NaCl in sweat.

In GIT & lungs : There is inability to transport chloride outside cells → chloride collects intracellularly → increased absorption of Na+ & water → collection of hyperviscous mucus that blocks passages that causes recurrent infections and inflammation → organ damage.

Other findings - Meconium Ileus, FTT, Exocrine Pancreatic insufficiency.

Diagnosis: Immunoreactive trypsinogen, Sweat chloride test (chloride value >=60 mmol/L is diagnostic), Gene testing.

→ Pseudomonas is the most common cause of Pneumonia in patients with Cystic Fibrosis in adults (Staph Aureus children).

3. A child has recurrent bone fractures and bluish sclera ?

Osteogenesis imperfecta

Inheritance : Autosomal dominant

PATHOGENESIS: Mutation in COL1A1 & COL1A2 →decreased hydrogen and disulfide bonds between type 1 preprocollagen molecules → poor triple helix formation, therefore poorly formed type 1 collagen.

CLINICAL FEATURES : BITE

Bone = Recurrent fractures, brittle bones, bow legs

TIP : multiple in utero fractures = Osteogenesis imperfecta.

Eye (I) = blue sclera (due to the underlying choroidal veins being visible through the thin sclera, not pigmentation)

Teeth = brittle, discolored teeth (dentinogenesis imperfecta)

Ear = Hearing loss (conductive)

Treatment : Bisphosphonates : Decrease the risk of fractures.

Differential :

Child abuse: Both conditions can have fractures. Whenever you get confused, look for specific features, eg : for osteogenesis imperfecta look for blue sclera, hearing loss & for child abuse look for Posterior rib fractures, retinal hemorrhage, history being inconsistent with the clinical presentation.

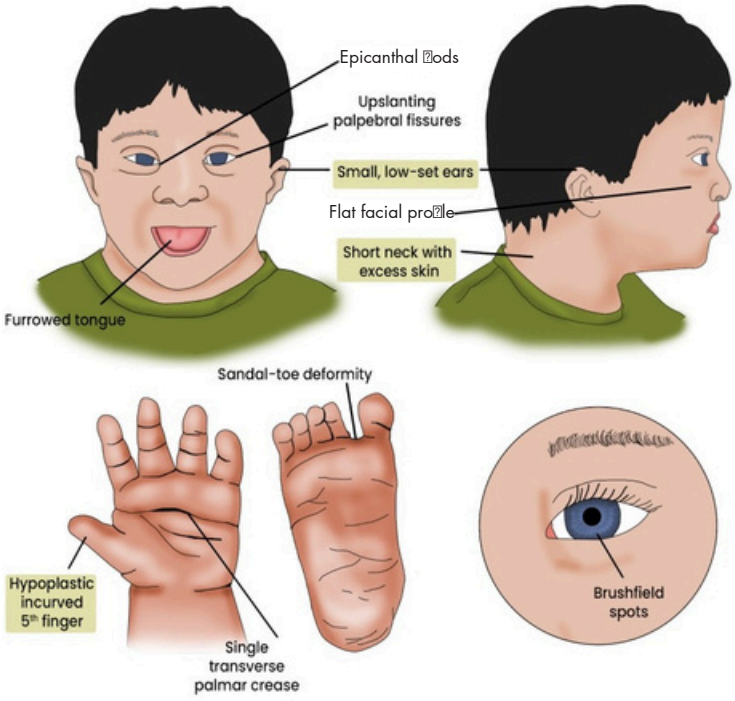
4.A pregnant female comes for routine follow up. Her USG shows increased nuchal translucency. Serum beta HCG is high and AFP is low ?

Down syndrome (Trisomy 21).

Most common cause : meiotic nondisjunction [during meiosis I > meiosis II]

Another possible cause : Robertsonian translocation between chromosome 14 & 21.

Here are all the features of down syndrome.



They can give any combination in the question.

How can they describe brushfield spots : whitish spots at the periphery of iris.

Important associations that you should be knowing for down syndrome : Hirschsprung disease, duodenal atresia, increased risk of AML & ALL,early onset Alzheimers.

USG findings:increased nuchal translucency & midface or nasal bone hypoplasia.

Importantarrows:

⬇ AFP ⬆ betaHCG ⬇ Estriol ⬆ inhibin A⬇ PAPP- A

Conformation : Karyotyping.

TIP for Edwardsyndrome : Trisomy 18 → at 18 we are adults and start doing everything bad that makes us DOWN, so all the arrows are downforEdward syndrome.

5. A patient has sudden chest pain radiating back. ECHO shows aortic dissection. He has a tall stature, long limbs & upward lens dislocation ?

Marfan syndrome

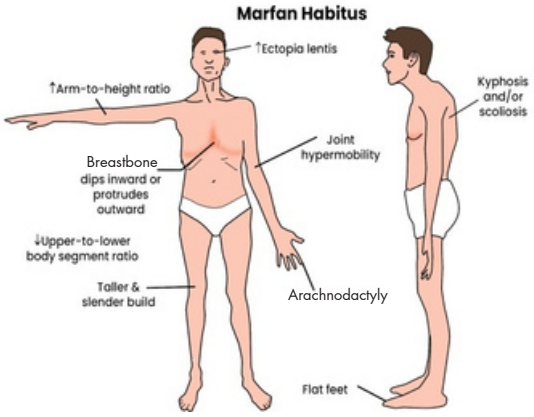
Inheritance: autosomal dominant with variable expression.

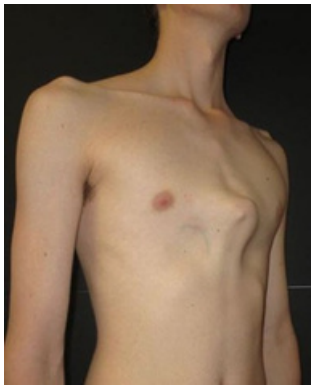
Gene mutation ? = FBN 1 gene → leads to defective fibrillin.

Type of mutation ? = missense mutation > splice site mutation.

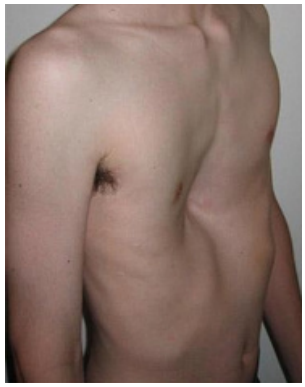
Normal function of fibrillin ? = forms a sheath around elastin.

Here are all the possible physical features of marfan syndrome (Marfanoid habitus) :





Sternum growing outward → known as Pectus carinatum (pigeon chest).



Sternum growing inward → known as Pectus excavatum.

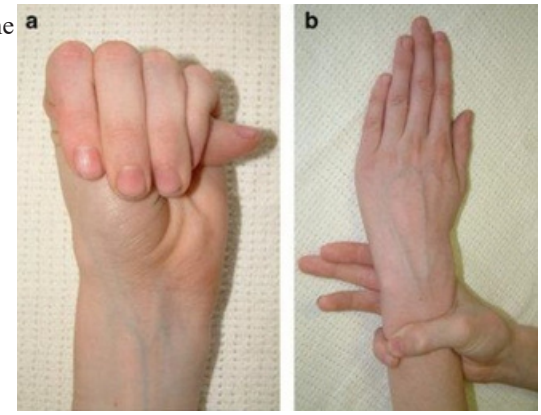


Image a → Thumb sign : Thumb protrudes beyond the ulnar border of the hand, when the patient is asked to make a fist.

Image b → Wrist sign : The little finger and thumb overlaps when the patient is asked to clasp the wrist of the opposite hand.

Important associations that you should know :

- Increased risk of mitral valve prolapse.
- Upward & outward lens dislocation (marfan = fan is up).
  Increased risk of spontaneous pneumothorax (sudden chest pain,
- Decreased chest expansion, hyperresonance on percussion).
- Increased risk of aortic root dilation/ aneurysm → leading to aneurysm rupture or aortic dissection.

Reason for aortic issues = cystic medial necrosis due weak elastin in aorta.

Most common cause of death in marfan syndrome = Aortic dissection.

Intellect in marfan syndrome : Normal.

Differentials of Marfanoid habitus :

Homocystinuria : look for thrombosis, downward lens dislocation, intellectual disability, etc.

MEN 2 B : also have medullary thyroid cancer & pheochromocytoma.

6. A patient has recurrent joint dislocations, and his skin is hyperextensible & bruise easily?

Ehler danlos syndrome

Etiology : Mutation in genes responsible for synthesis and processing of collagen (especially type 3 collagen) such as COL3A1 gene.

Can also be caused by **procollagen peptidase deficiency.**

Clinical Features :

- Hyperextensible, easily bruised skin.
- Hypermobile joints
- Poor wound healing
- Increased risk of mitral valve prolapse
- Increased risk of aortic issues (aortic root dilation, dissection).
- **Increased risk of berry aneurysm → subarachnoid hemorrhage.**

TIP : If you get a stethoscope question in your exam in a patient with Marfan syndrome or Ehler danlos syndrome, think of Mitral valve prolapse or Aortic regurgitation.***

Extra edge: Classic type Ehler danlos: mutation- COL5A1/COL5A2 and cardiac presentation is uncommon.
Vascular type(type IV) - more associated with COL3A1 and risk of arterial rupture.

7. A patient has a cherry red spot on the macula, no hepatosplenomegaly and hyperacusis?

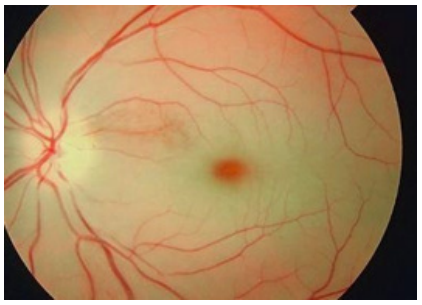
Tay Sachs disease

Inheritance : Autosomal recessive

What's deficient = Hexosaminidase A

What's accumulated substance = GM2 ganglioside

TIP : Just remember **Cherry red spot + NO hepatosplenomegaly = Tay sachs.**



Differentials of cherry red spot :

Niemann pick : **Cherry red spot + Hepatosplenomegaly** (sphingomyelinase deficient, sphingomyelin accumulation)

Central retinal artery occlusion : has painless, monocular vision loss, also look for history of atherosclerosis, atrial fibrillation, stroke or transient ischemic attack.

8. A patient has extreme bone pain, hepatosplenomegaly, and pancytopenia ?

Gaucher disease

Inheritance - Autosomal recessive

What's deficient = Glucocerebrosidase (B-glucosidase)

What's accumulated substance = Glucocerebroside

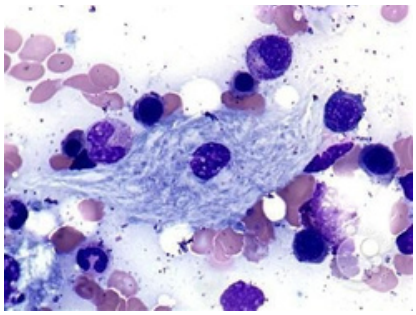
Features :

Extreme bone pain (bone crisis)

Hepatosplenomegaly

Pancytopenia

Avascular necrosis of femur (hip pain, limited internal rotation).



Histology important :

Lipid laden macrophages that look like a crumpled tissue paper.

Do we have a Treatment ? : yes, recombinant glucocerebrosidase.

9.A patient has burning pain in hands & feet, keratotic papules that sometimes bleed, and he has decreased sweating as compared to his peers while he plays weekly basketball tournaments ?

Fabry disease

Inheritance : X linked recessive

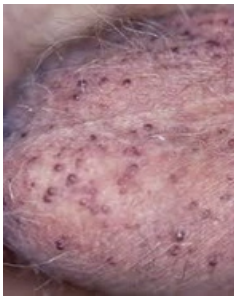
What's deficient = Alpha galactosidase A

What's accumulated = Ceramide trihexoside.

Features :

Classic triad of : Peripheral neuropathy, Angiokeratomas (keratotic lesions that may be pruritic or painful or may bleed sometimes) and Hypohidrosis.

Complications : Renal failure, cardiomyopathy, cataract.



(Angiokeratoma)

10. A newborn has musty body odor, hypopigmented skin and seizure ?

Phenylketonuria

Inheritance = Autosomal recessive.

Etiology : Either deficiency of phenylalanine hydroxylase or deficiency of Tetrahydrobiopterin (BH4, a cofactor for phenylalanine hydroxylase).
- BH4 is also a cofactor of tryptophan hydroxylase, so deficiency can cause decreased synthesis of catecholamines (decreased tyrosine by phenylalanine hydroxylase) and decreased synthesis of neurotransmitters (decreased serotonin by tryptophan hydroxylase).

Pathogenesis : Whatever the etiology maybe, phenylalanine won't convert into tyrosine → ⬆ levels of phenylalanine → ⬆ phenyl ketones in urine.

Features :

Intellectual disability

Seizures

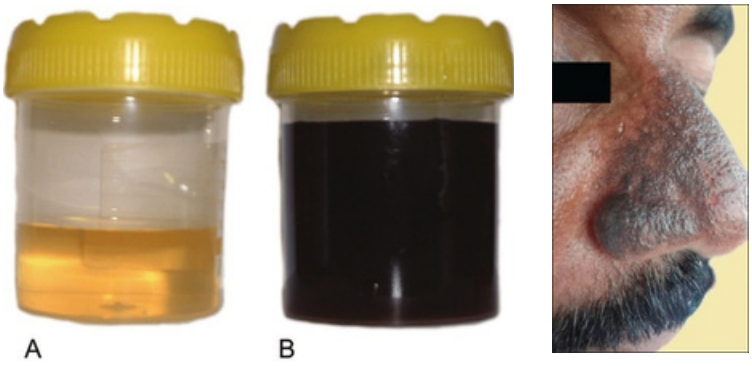
Eczema Musty body odor

(because phenylalanine is an aromatic amino acid).

Hypopigmented skin and pale hair (as tyrosine is low, it won't convert to melanin).
If a mother has PKU, findings in the neonate will be :
Growth restriction
Microcephaly
Abnormal facial features
Congenital heart defects
Intellectual disability
Diagnosis : neonatal screening
Treatment : Diet ⬇ in phenylalanine & ⬆ in tyrosine, such as chicken, fish, etc.
Avoid aspartame (an artificial sweetener that contains phenylalanine).

11. A child has joint pain and mother says his sweat appears blackish ?

Alkaptonuria
Inheritance = Autosomal recessive
Pathogenesis : Deficiency of homogentisate oxidase → homogentisic acid won't convert to maleylacetoacetate & fumarate → homogentisic acid starts accumulating everywhere and gives symptoms.
Features:
Bluish black discoloration of cartilage (such as ear), tendons, sclera.
Urine gets dark when exposed to air. Sweat may appear dark.
Arthralgias, arthritis (due to deposition of homogentisic acid in articular cartilage).



Treatment: Diet low in tyrosine and phenylalanine to reduce their conversion to homogentisic acid.

12. A child has poor feeding smd mother says his urine smells like burnt sugar ?

Maple syrup urine disease
Inheritance = Autosomal recessive
Pathogenesis : **Deficiency of branched chain alpha ketoacid dehydrogenase** → poor degradation of branched chain amino acids (leucine, isoleucine, valine) → increased alpha keto acids.
Features : Multiple symptoms like poor feeding, vomiting, etc. but one thing that questions always mention is : **Burnt sugar or Maple syrup like the smell of urine.**
Complication : Neurological decline
Diagnosis : Part of newborn screening
Increased serum alpha keto acids & branched chain amino acids.
Treatment : Restrict valine, leucine & isoleucine in diet
Give **Thiamine supplementation** (acts as a cofactor for branched chain alpha ketoacid dehydrogenase).

12. A female fasted for 6 days for a study. She was allowed to have water in the morning. What metabolic processes will be deactivated ?

- Glycolysis : Deactivated due to depletion of glucose stores.
- Glycogenesis : As there is not enough glucose to store as glycogen.
- Lipogenesis : Fatty acid synthesis from acetyl- CoA and glucose is reduced due to less glucose available.

Important arrows for fasting :

⬇ Fructose - 2,6 - bisphosphonate production

⬇ Phosphofructokinase 2 activity

FED STATE	FAST STATE
Anabolic	Catabolic
Glycogen synthesis	Glycogenolysis
Cholestrol synthesis	Gluconeogensis
FA synthesis	FA oxidation
Glycolysis	

13. A patient has a problem with vesicular trafficking proteins. What vesicular trafficking proteins do we have ?

I cell diseases

- Clathrin : involved in receptor mediated endocytosis and transports vesicles from golgi to endosomes and lysosomes (remember the problem is in golgi apparatus)
- Coat Protein Complex I (COP I): retrogradely transport vesicles from golgi to endoplasmic reticulum.
- Coat Protein Complex (COP II) : anterogradely transport vesicles from endoplasmic reticulum to golgi.
- I-cell disease is due to **defective N-acetylglucosaminyl-1-phosphotransferase,** leading to failure to add **mannose-6-phosphate** to lysosomal enzymes.

14. A man consumed lots of beer for 2 weeks and now he has confusion & ophthalmoplegia. If the deficient vitamin is given, RBCs will show increased activity of what enzyme ?

Transketolase

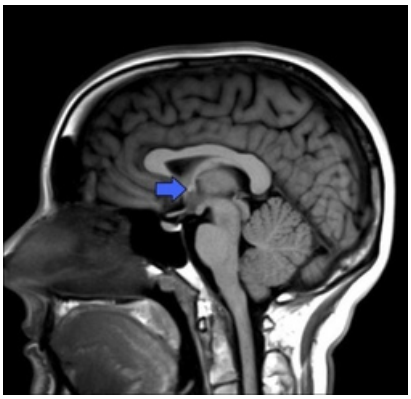
Active component of vitamin B1 = Thiamine pyrophosphate (TPP).

TPP acts as a cofactor for :

- Pyruvate dehydrogenase (convert pyruvate to acetyl- CoA).
- Alpha ketoglutarate dehydrogenase (convert alpha ketoglutarate to succinyl CoA).
- Branched chain amino acid dehydrogenase
- Transketolase

What does B1 deficiency lead to -

- Dry beri beri : Peripheral neuropathy
- Wet beri beri : High output heart failure (S3, crackles, etc).
- Wernicke encephalopathy : Confusion, Ophthalmoplegia, ATaxia (COAT)
- Korsakoff syndrome : Confabulation, Amnesia (retrograde & anterograde), Personality changes (CAP). Auterograde > retrograde



(Mamillary body)

What happens in Wernicke Korsakoff syndrome ? = damage to mammillary bodies & medial dorsal nucleus of thalamus.

Diagnosis : Increase in RBC transketolase activity after B1 supplementation.

Treatment : Thiamine before glucose.

15. A patient develops nausea, bloating, abdominal pain, diarrhea, vomiting & low blood sugar after ingesting fructose. What enzyme is deficient ?

Aldolase B

General things about Fructose : It is absorbed by Facilitated diffusion by GLUT 5 (Five).

Fructose disorders :

Essential fructosuria : Fructokinase deficiency → asymptomatic/ incidentally found.

Hereditary fructose intolerance : aldolase B deficiency (symptomatic).

Possible symptoms (other than the ones given in question) :

Jaundice, Cirrhosis.

Symptoms begin when the child is weaned off breast milk and starts consuming food that contains sucrose like fruit juice, honey etc..

Why does aldolase B deficiency give symptoms? = due to accumulation of Fructose - 1 - phosphate.

Diagnosis : Reducing sugars in urine.
Treatment : No fructose, sucrose or sorbitol.

TIP : Reducing substances in urine = fructose or galactose disorders.

16. A patient with colon cancer undergoes a genetic study that shows -DTEA DTEA DTEA. What is the principle of the enzyme in this pathology ?

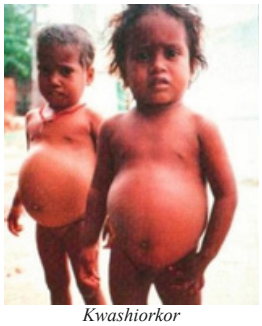
Gene duplication.
The enzyme is Telomerase.
What is telomerase ? A reverse transcriptase (RNA dependent DNA polymerase).
What does it do ? Adds DNA (TTAGGG) to 3' end of chromosomes → this maintains & lengthens the telomeres → prevention of chromosome shortening.
When does telomerase increase ? Cancer.
When does telomerase decrease ? Aging.

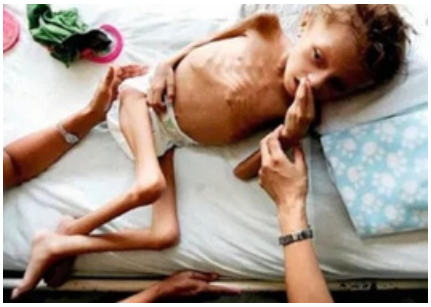
17. A child from Africa has bilateral pitting edema in lower limbs, muscle atrophy, thin dry skin, hepatomegaly ?

Kwashiorkor
Pathogenesis : A protein energy malnutrition → low protein leads to low oncotic pressure → edema.
Decreased apolipoprotein synthesis → fat deposition in liver → hepatomegaly.

Features:

- Bilateral pitting edema
- Hepatomegaly → distended abdomen.
- Skin changes (thin dry skin, hyperpigmentation)
- Dry hair Muscle atrophy


Kwashiorkor


Marasmus

Differential :
Marasmus : Loss of all major nutrients . Can be confused with Kwashiorkor, but it has **extreme muscle wasting and no edema.**

18. A patient says every time he starts exercising he gets severe muscle pain/ cramps that resolve after a few minutes?

McArdle disease
Enzyme deficient = Glycogen phosphorylase or myophosphorylase.
Consequence = Too much glycogen in skeletal muscles which cannot be broken down.
Other features :

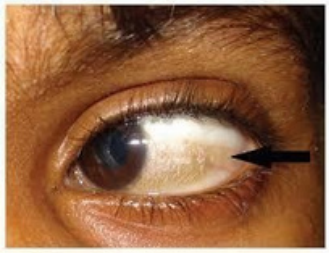
- Dark urine (myoglobinuria) during exercise.
- **No increase in venous lactate** (but normal increase in ammonia level) during exercise.

Complication = Arrhythmias

19. A patient has dry skin, hepatomegaly. He recently received treatment for acne. What could be the cause ?

Vitamin A toxicity
Important points about Vitamin A :

- Important for Vision.
- Important for cellular differentiation or maintenance of specialized tissue.
- Prevents squamous metaplasia → prevention of skin cancer.
- Deficiency : night vision changes, dry skin & eyes, bitot spots (due to squamous metaplasia of conjunctiva), corneal degradation/ softening (keratomalacia).



Toxicity :
Vitamin A excess + nausea, vomiting, vision changes = Pseudotumor cerebri.
Alopecia, dry skin, hepatotoxicity.

Pharmacologic uses :
All trans retinoic acid used in Acute promyelocytic leukemia (induces differentiation of immature cells into mature neutrophils).

Vitamin A supplement is given in Measles.
Isotretinoin (derivative of Vitamin A) is used to treat severe acne.
How isotretinoin works ? = A transcription factor that inhibits sebum production.
Test to do before prescribing Isotretinoin ? = **Pregnancy test** as it is highly teratogenic (cleft lip/ palate, microtia,thymic defects, etc).
How isotretinoin causes teratogenicity ? = Disrupts HOX gene expression.

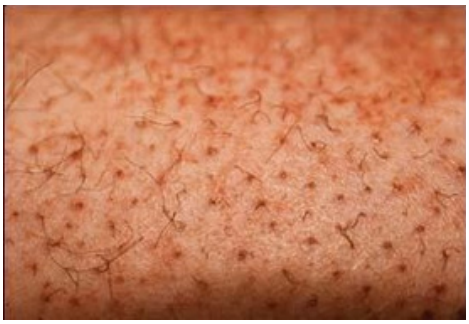
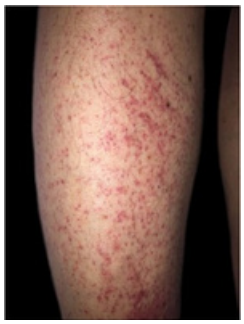
20.A patient who mainly consumes oatmeal and tea has petechiae, follicular keratosis. What is the major function of the deficient vitamin ?

- Hydroxylation of proline & lysine in collagen synthesis.

Diagnosis : Vitamin C deficiency
Another important function of Vitamin C : Iron resorption by converting Fe 3+ to Fe 2+.
Vitamin C deficiency leads to :

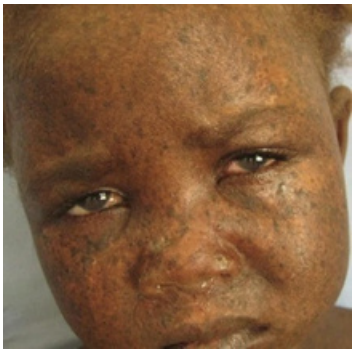
Scurvy :-

- Follicular hyperkeratosis
- Perifollicular & subperiosteal hemorrhage (can lead to difficulty walking).
- Corkscrew or coiled hair
- Swollen, red, bleeding gums
- Petechiae, easy bruising.
- Impaired wound healing.
- Arthralgia
- Hemarthrosis

Question usually refers to a malnourished patient (chronic alcoholic, those on tea & toast diet).
Any therapeutic use of vitamin C ? = Methemoglobinemia.
Labs/ arrows : Anemia
Normal platelet count (despite petechiae & mucosal bleeding).
Normal PT, PTT (bleeding is due to vascular fragility, not platelet or coagulation factor deficiency).
⬆ Bleeding time.
About vitamin C toxicity : just remember it can lead to Calcium oxalate stones.
Other causes of Hemarthrosis : ACL tear , Hemophilia.

21.A patient develops blistering skin lesions after minimal exposure to sunlight. Genetic study reveals defect in repair of damaged pyrimidine dimers caused by UV exposure ?



Xeroderma pigmentosum.
Etiology : Nucleotide excision repair defect.
Features : Photosensitivity, hyperpigmentation, skin cancer.
Other organs involved : Ocular (photophobia, blindness) > Nervous system (ataxia, hearing loss, etc.).
What happens in Nucleotide excision repair ? Endonucleases remove the damaged DNA dimers → DNA polymerase & ligase fill and reseal the gap.
When does Nucleotide excision repair occur ? During the **G1 phase.**

Differential :
Porphyria cutaneatarda(PCT): Also have blistering skin lesions after sun exposure. If the question wants to give PCT as the answer, they will give some association with liver, exacerbation with alcohol consumption.

22. A newborn has spina bifida. The vitamin that the mother didn't take during pregnancy plays a role in which reactions?

Methylation reactions
Vitamin B9 deficiency

Another function : makes nitrogenous bases in DNA & RNA.

Where is B9 absorbed ? Jejunum (Iron in duodenum, Folic acid in jejunum and B12 in ileum).

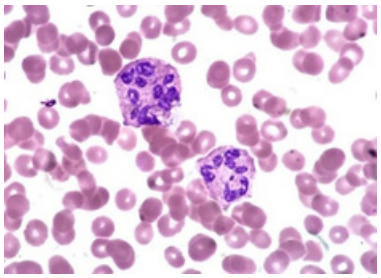
Etiology of deficiency :

- Mother not taking B9 during pregnancy.
- Alcoholics
- Drugs (phenytoin, trimethoprim, methotrexate, valproate, carbamazepine) ***

Features :

Megaloblastic anemia (signs of anemia will be present).

Teratogen : neural tube defects.



Labs :

MCV >100 fL

⬆ homocysteine & Normal methylmalonic acid levels.

Blood smear : Hypersegmented neutrophils.

Dihydro Folate reductase inhibitors :

- Trimethoprim
- Pyrimethamine
- Methotrexate

Differential :

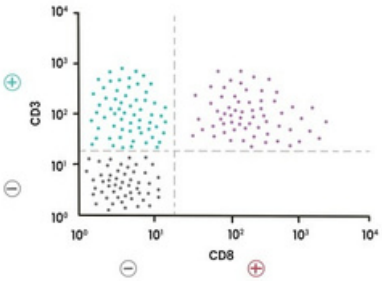
Vitamin B 12 deficiency : Have ⬆ homocysteine & ⬆ methylmalonic acid.

Side note : A patient with megaloblastic anemia + neurological symptoms + vitamin B9 given + anemia resolves but neurologic symptoms persist or worsen → Vitamin B 12 deficiency.

23. The following diagram is given :

Which lab test is used to determine these CD markers ?

Always remember, **CD markers = Flow cytometry.**

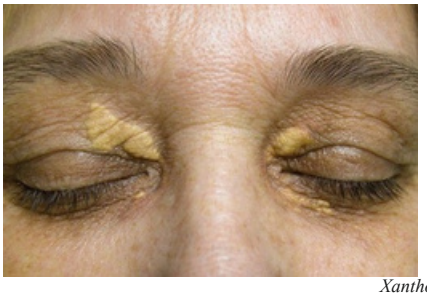


24. A blood sample is drawn and there is white stuff on blood in the test tube ?

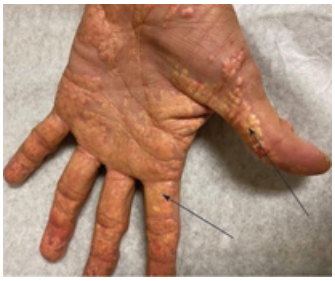
Increased triglycerides in blood.

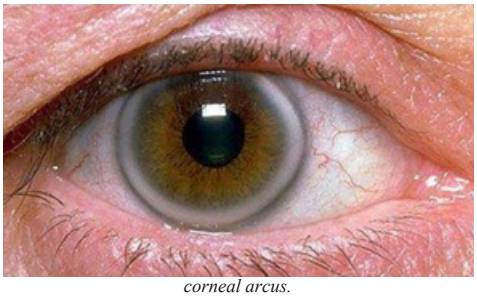
Familial hypercholesterolemia :

Type	Pathogenesis	Features
Type I- Hyperchylomicronemia	Deficiency of apo C2 or lipoprotein lipase → ⬆ chylomicrons, triglycerides.	Pancreatitis Creamy / whitish layer over blood sample. xanthoma.
Type II- Hypercholesmlemia	Absent or deficient LDL receptors or Apo B → ⬆ LDL, triglycerides, cholesterol.	Cholesterol = ~300 - 600 mg/dL if deficiency of LDL receptors. Cholesterol = >700 mg/dL if complete absence of LDL receptors. Increased risk of accelerated atherosclerosis (can cause young onset Myocardial infarction). Corneal arcus Tendinous xanthoma
Type III- Dysbetalipoproteinemia	Defective Apo E3 or E4	Premature atherosclerosis. Palmar xanthoma
Type IV - Hypertriglyceridemia	Overproduction of VLDL → ⬆ VLDL & triglycerides.	Pancreatitis Usually in **diabetics.**



Xanthomas





corneal arcus.