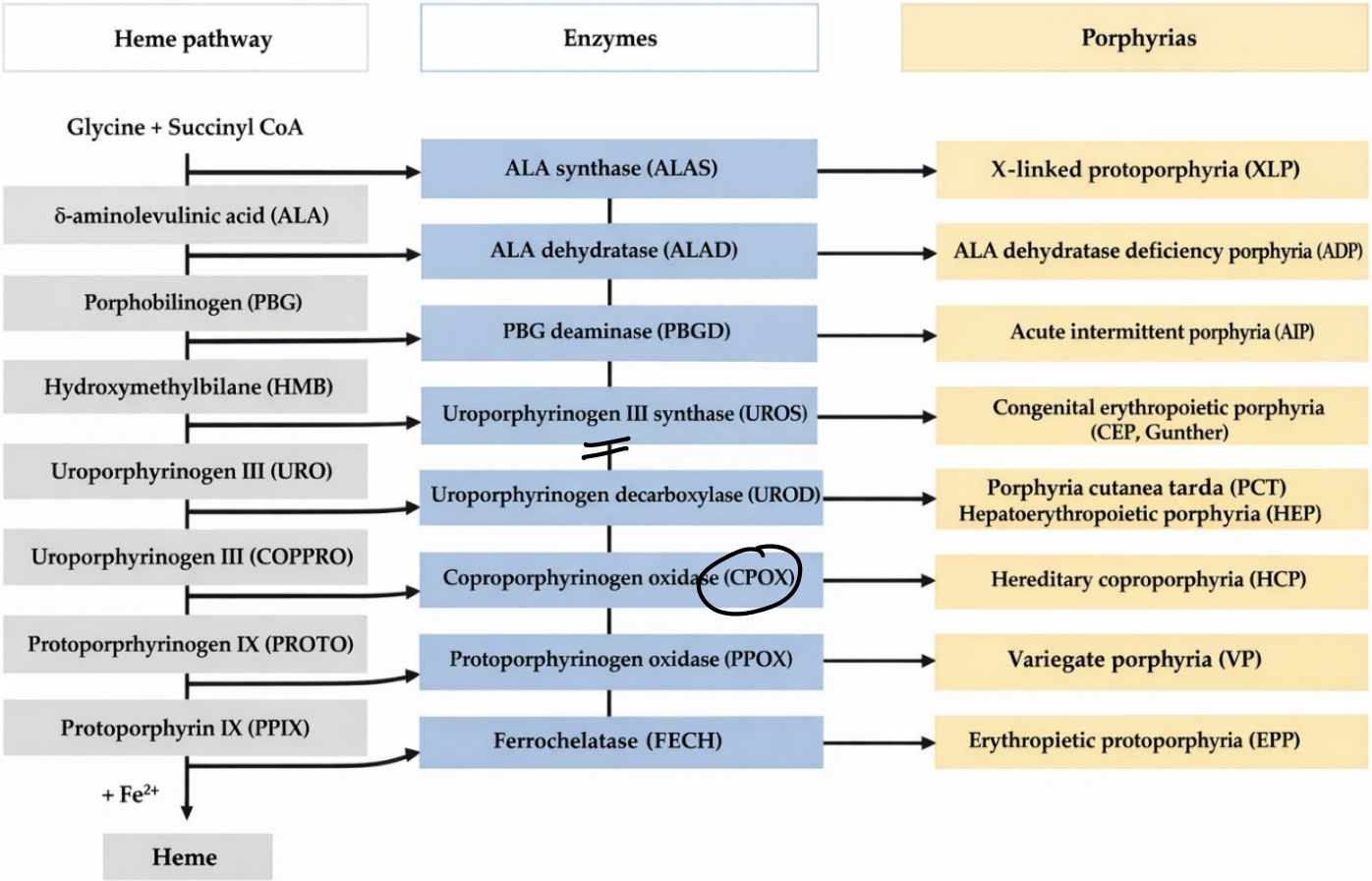
BIOCHEMISTRY


Heme pathway
Enzymes
Porphyrias
Glycine + Succinyl CoA
δ-aminolevulinic acid (ALA)
Porphobilinogen (PBG)
Hydroxymethylbilane (HMB)
Uroporphyrinogen III (URO)
Uroporphyrinogen III (COPPRO)
Protoporprhyrinogen IX (PROTO)
Protoporphyrin IX (PPIX)
+ Fe2+
Heme
ALA synthase (ALAS)
ALA dehydratase (ALAD)
PBG deaminase (PBGD)
Uroporphyrinogen III synthase (UROS)
Uroporphyrinogen decarboxylase (UROD)
Coproporphyrinogen oxidase (CPOX)
Protoporphyrinogen oxidase (PPOX)
Ferrochelatase (FECH)
X-linked protoporphyria (XLP)
ALA dehydratase deficiency porphyria (ADP)
Acute intermittent porphyria (AIP)
Congenital erythropoietic porphyria (CEP, Gunther)
Porphyria cutanea tarda (PCT)
Hepatoerythropoietic porphyria (HEP)
Hereditary coproporphyria (HCP)
Variegate porphyria (VP)
Erythropietic protoporphyria (EPP)

AIP	Acute Abdomen, insanity, P. neuropathy PBG ↑ PHOTOSENSITIVITY ⊖ OCP Triggers: Drugs (antiepileptics), alcohol, fasting Treatment: Hemin and glucose
CEP GUNTHER	 VROS child, PTS ++ Red Teeth, Red urine
PCT **PUDCT**	VROD PTS, HYPERTRICHOSIS, TEA color urine

HCP	Coproporphyrinogen oxidase Step: Converts coproporphyrinogen III → protoporphyrinogen IX (mitochondria) Neurovisceral + Photosensitivity
VP	Protoporphyrinogen oxidase Step: Converts protoporphyrinogen IX → protoporphyrin IX (mitochondria) Neurovisceral + Photosensitivity South African patient
EPP **Extreme pain on photo**	Ferro chelatase Converts protoporphyrin IX → heme (adds Fe^{2+}) Pure photosensitivity minus blistering

Neurovisceral

1. A 18-year-old girl presents with recurrent severe abdominal pain, vomiting, and anxiety. She also complains of blistering lesions over sun-exposed areas. Her urine becomes dark on standing. There is no significant drug history. Which of the following is the most likely diagnosis?

a. Acute intermittent porphyria PTS⊖

b. Porphyria cutanea tarda PUDCT UROD

c. Hereditary coproporphyria → CPOX

d. Erythropoietic protoporphyria PTS+

RP

2. A 20-year-old man presents with progressive night blindness, peripheral neuropathy, and anosmia. Examination reveals retinitis pigmentosa and cerebellar ataxia. What is the most likely diagnosis?

a. Zellweger syndrome

b. Refsum disease

c. Krabbe disease

d. Metachromatic leukodystrophy

aryl sulfatase B *

* Cant see — RP
* " smell — Anosmia
* " feel — P. neuropathy
RP ataxia

PCoA Hydroxylase ⊖

zero peroxisome
non functional "

VLCFA +
phytanic acid +
bile acid derivatives +

XLD : ADL

ABCD1 Transporter
addison

Refsum disease

- Can’t see, Smell and can’t feel
- Defective α-oxidation of phytanic acid
- Phytanic acid derived from green leafy vegetables, fish and animal fat
- Autosomal recessive, PHYH deficiency

Zellweger syndrome

- Zero Peroxisomes due to PEX gene
- VLCFA accumulation leading to floppy baby
- Neonate with RP and Cerebellar ataxia

Adrenoleukodystrophy

- X-linked disorder (boys)
- Defect: VLCFA accumulation (ABCD1 gene)
- Affects brain + adrenal (demyelination + Addison)
- MRI: posterior (parieto-occipital) white matter lesions
- Treatment: Lorenzo oil + early HSCT

3. A 45-year-old man presents with recurrent blistering lesions over the dorsum of hands, especially after sun exposure. He also reports increased facial hair and darkening of urine on standing. He has a history of alcohol use. On examination, there are fragile skin lesions with erosions and crusting over sun-exposed areas. What is the most likely underlying enzyme deficiency?

a. Uroporphyrinogen decarboxylase

b. Porphobilinogen deaminase

c. ALA synthase

d. Ferrochelatase

UROD

PUDCT

F : Fasting

A : alcohol

C : Cycles Menses

T : Toxin/drugs : OCP, AED

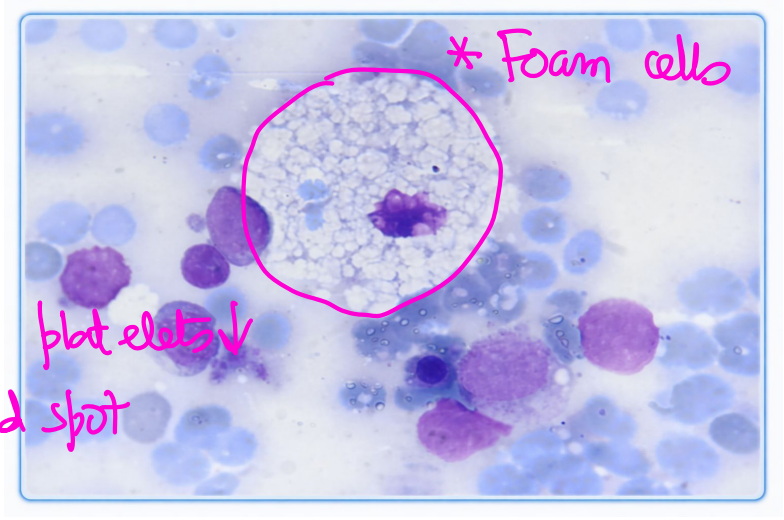
4. A 3-year-old child presents with progressive abdominal distension and hepatosplenomegaly. On examination, there is developmental delay and hypotonia. Bone marrow biopsy is shown below. Fundoscopy reveals a cherry-red spot in the macula. What is the most likely diagnosis? *

a. Gaucher disease

b. Niemann-Pick disease

c. Tay-Sachs disease

d. Fabry disease



G: HSM + Bone pain, anemia, platelets↓

NP: HSM + ND + cherry Red spot

T: HSM⊖ ND + " "

F: neuropathic pain, XLR

Gaucher disease	Beta -glucocerebrosidase	A: Anemia B: Bone pain C: Crumpled Tissue appN D: Distened abdo: Spleen ++ E: ERLENMEYER FLASK ERT: IMIGLUCERASE
Niemann-pick disease	Sphingomyelinase	
Tay -sachs disease	Hexosaminidase A	
Fabry disease	α-galactosidase A	

5. A 6-month-old infant presents with progressive neurodegeneration, exaggerated startle response, and developmental regression. Fundoscopy shows a cherry-red spot. On examination, there is hepatosplenomegaly. Diagnosis is?

a. Tay Sachs disease

b. Fabry disease

c. Sandhoff disease

d. Gaucher disease

Ashkenazi Jews ND + cherry Red spot

ND + Cherry Red spot + HSM

Feature	Tay-Sachs disease	Sandhoff disease
Enzyme defect	Hexosaminidase -A ↓	Hexosaminidase A+B↓
Substrate	GM2 ganglioside ↑	GM2 ganglioside ↑
Ethnicity	Ashkenazi jews	No specific (more common in non-jew/ Indian populations)
Onset	Infant (3-6 months)	Infant (similar)
Neuro features	Severe neurodegeneration, hyperacusis	Same
Cherry -red spot	Present	Present
Hepatosplenomegaly (HSM)	Absent	Present
Histology	Foam cells, lysosomes with ganglioside	Similar
IOC (investigation of choice)	Enzyme assay → Hex A ↓	Enzyme assay →Hex A+B↓
Treatment	No cure	No cure

GM1 Gangliosidosis	GM1 ganglioside *	β-galactosidase
GM2 — Tay Sachs	GM2 ganglioside ✓	Hexosaminidase A
GM2 — Sandhoff	GM2 ganglioside ✓	Hexosaminidase A + B

6. A 4-month-old infant presents with poor feeding, hypotonia and progressive respiratory difficulty. CXR shows CT ratio of 0.7. Echocardiography shows hypertrophic cardiomyopathy. Which of the following investigation should be done in this patient?

a. Urine spectrophotometry

b. Muscle biopsy with PAS staining

c. Cardiac biopsy with PAS-D staining

d. Liver biopsy with PAS Staining

POMPE
acid maltase
floppy baby + HCM
Skeletal muscle
Cardiac muscle
glycogen +

7. A 22-year-old man presents with exercise intolerance, early fatigue, and muscle cramps. He reports a "second wind" phenomenon and occasional dark-colored urine after exertion. An ischemic forearm exercise test is performed. Which of the following results is most consistent with this condition?

a. Increased lactate with normal ammonia

b. Normal lactate with increased ammonia

c. Increased lactate with increased ammonia

d. Decreased lactate with decreased ammonia

* Glycogen in muscle ≠ ATP
* fat → FFA → β oxidation → ATP
+

decreased

(n): Glycogen: lactate ++
NH_3 ++
EXERCISE

AMP → IMP + NH_3

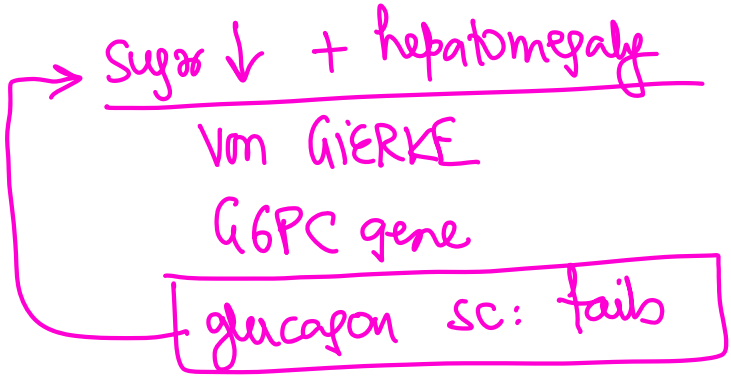
- ✓ Defect in muscle glycogen phosphorylase
- ✓ No glycolysis explains no lactate rise
- ✓ Muscle now looks for alternative source of energy
- ✓ It causes Purine metabolism and ammonia rises

2 ADP → ATP + AMP (via adenylate kinase)

AMP → IMP + NH_3 (via AMP deaminase)

8. A 1-year-old child presents with protuberant abdomen, recurrent episodes of severe fasting hypoglycemia, and irritability. On examination, there is hepatomegaly. Laboratory findings show elevated lactate, triglycerides, and uric acid. The following disease occurs due to defect on which chromosome

a. Chromosome 17
b. Chromosome 18
c. Chromosome 19
d. Chromosome 20



9. A 1-month-old infant presents with difficult to control seizures, and a musty odor of urine/diapers. On examination, the child has hypopigmented skin and hair. Which of the following metabolites is responsible for the neurological manifestations in this condition?

a. Tyrosine

b. Phenylacetate

c. Phenylpyruvate

d. Homogentisic acid

P.K.U

musty

* phenylacetate : URINE +
* " pyruvate : ++
* Melanin ↓ : blond hair
  blue iris

10. A 3-month-old infant is diagnosed with Phenylketonuria on newborn screening. Despite dietary restriction of phenylalanine, serum phenylalanine levels remain elevated. A trial of tetrahydrobiopterin (BH4) leads to a significant reduction in phenylalanine levels. Which of the following best explains this response?

a. Complete absence of phenylalanine hydroxylase

b. Partial deficiency of phenylalanine hydroxylase

c. Increased degradation of tyrosine

d. Defect in homogentisate oxidase

BH4 cofactor for PAH

SAPROPTERIN

11. A 40-year-old man presents with chronic low back pain and stiffness. He reports that his urine turns dark on standing. On examination, there is bluish-black pigmentation of ear cartilage and reduced lumbar flexion. Which of the following vitamins can be used in this condition?

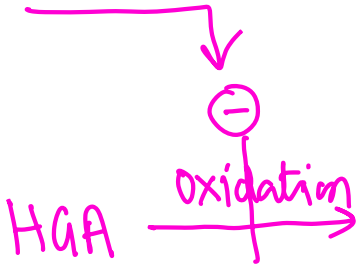
a. Vitamin A

b. Vitamin B12

c. Vitamin C

d. Vitamin D

* OCHRONOSIS : ↓
* SCHOBER TEST : OA spine
* TURNS black on air exposure



12. A 38-year-old man presents with chronic low back pain, bluish-black pigmentation of ear cartilage, and urine that darkens on standing. He is started on nitisinone therapy. What is the mechanism of action of this drug?

a. Increases conversion of homogentisic acid to maleylacetoacetate

b. Inhibits homogentisate oxidase enzyme

c. Inhibits 4-hydroxyphenylpyruvate dioxygenase

d. Enhances renal excretion of homogentisic acid

ALKAPTONURIA

⊖ 4-HPP dioxygenase

↓

HGA ↓

NITISINONE →

13. A 10-year-old child from village presents with school absenteeism, photosensitive rash over face, ears and neck with intermittent diarrheal episodes and episodes of ataxia. Work up shows aminoaciduria. What is the most likely underlying defect?

a. Pellagra

b. Hartnup disease

c. Maple syrup urine disease

d. Alkaptonuria

DERMATITIS ← NIACIN↓

DIARRHEA ←

neutral AA

Tx defective

SLC 6A19

ATAXIA

* Jejunum
* Renal Tubules

Tryptophan cannot be absorbed

Sepsis ?

14. A 2-day-old male neonate presents with poor feeding, vomiting, lethargy, and seizures. Laboratory evaluation reveals marked hyperammonemia. Urine examination shows increased orotic acid. Which of the following enzymes is most likely deficient?

a. Carbamoyl phosphate synthetase I

b. Ornithine transcarbamylase

c. Argininosuccinate synthetase

d. Argininosuccinate lyase

NH3 + CO2
↓ CPS-I (mitochondria, N-acetylglutamate cofactor)
Carbamoyl phosphate → spills over cytoplasm
~~↓ OTC (mitochondria)~~ (−)
Citrulline → exits to cytoplasm
↓ ASS - Argininosuccinate synthetase (cytoplasm)
Argininosuccinate → + pyrimidine Metabolism
↓ ASL - Argininosuccinate lyase (cytoplasm)
Arginine + Fumarate → + ↑ OROTIC Acid
↓ Arginase (cytoplasm)
Urea + Ornithine
↓
Ornithine re-enters mitochondria → cycle continues

Enzyme	Key clue
CPS I	Hyperammonemia, normal orotic acid
OTC	Hyperammonemia, ↑orotic acid
Argininosuccinate synthetase	↑citrulline
Argininosuccinate lyase	↑ Argininosuccinate

15. A 3-day-old neonate, previously well, develops poor feeding, vomiting, lethargy, and seizures after initiation of feeds. On examination, the baby is tachypneic. Labs shows:
Normal blood glucose
Normal blood lactate
ABG: pH: 7.48, pC02= 30 mm Hg and HCO3 =20 meq/L
What is the most likely underlying diagnosis?

R. alkalosis

a. Neonatal sepsis — M. ACIDOSIS

b. Glycogen storage disease

c. Urea cycle disorder — $NH_3 \uparrow$: + R. centre : $CO_2 \downarrow$

d. Organic acidemia — M. ACIDOSIS

16. A 3-day-old neonate presents with poor feeding, vomiting, lethargy, and seizures. Laboratory evaluation reveals marked hyperammonemia with normal blood glucose and respiratory alkalosis. The child is started on a drug that combines with glycine to form hippurate, which is excreted in urine, thereby reducing ammonia levels. Which of the following is the drug used?

a. Lactulose

b. Sodium benzoate

c. N-acetylcysteine PCM Toxicity

d. Metronidazole

17. A 50-year-old man presents with fatigue, pallor, and tingling sensations in his feet. Laboratory evaluation shows megaloblastic anemia. Serum folate levels are normal, but vitamin B12 levels are low. Which of the following best explains the mechanism of anemia in this patient?

folate Trap

a. Increased conversion of THF to dihydrofolate

b. Trapping of folate as methyl-THF due to vitamin B12 deficiency

c. Decreased synthesis of folate from dietary sources

methyl THF → THF
B12 ↓

d. Increased degradation of folate in liver

18. A 12-year-old boy presents with multiple yellowish papules over the extensor surfaces and buttocks. He has a history of recurrent abdominal pain. Fundoscopy reveals lipemia retinalis. Lipid profile shows markedly elevated triglycerides, and lipid electrophoresis demonstrates a prominent chylomicron band. What is the most likely underlying defect?

PREMATURE CAD, TENDON XANTHOMA → ERUPTIVE XANTHOMAS

a. LDL receptor deficiency IIA

b. Lipoprotein lipase (LPL) deficiency → PANCREATITIS → TG ↑ CM ↑

c. Apo B-100 deficiency IIB

d. Apo E deficiency

3(III palmar xanthome

I, IV, V

Type	Lipoprotein ↑	Defect	Lipid↑	Clinical features	Buzz word
Type I	Chylomicrons	LPL deficiency / Apo C- III	TG↑↑	Pancreatitis, eruptive xanthomas	Creamy plasma
Type IIa	LDL	LDL receptor defect	Cholesterol ↑	Tendon xanthomas, premature CAD	Achilles xanthoma
Type IIb	LDL +VLDL	Apo B-100 ↑	Cholesterol + TG↑	Mixed hyperlipidemia	Combined
Type III	IDL	Apo E defect	Chol + TG↑	Palmar xanthomas	Palmar crease
Type IV	VLDL	Overproduction	TG↑	Pancreatitis risk	VLDL excess
Type V	VLDL + Chylomicrons	Mixed Defect	TG↑↑	Pancreatitis eruptive xanthomas	Combo type

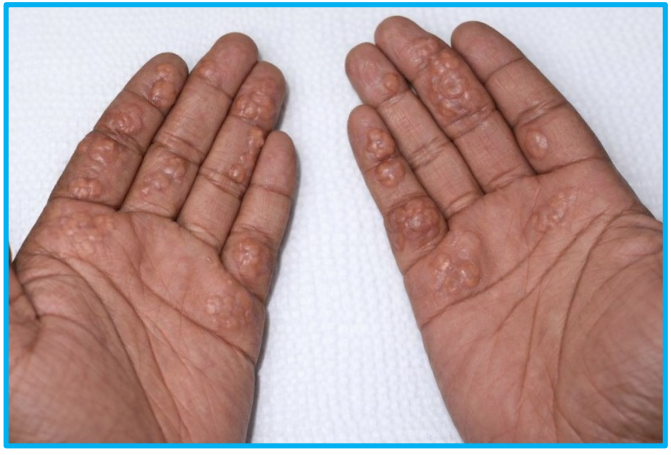
19. The following xanthomas are seen in which type of hyperlipoproteinemia?

a. Type I

b. Type II

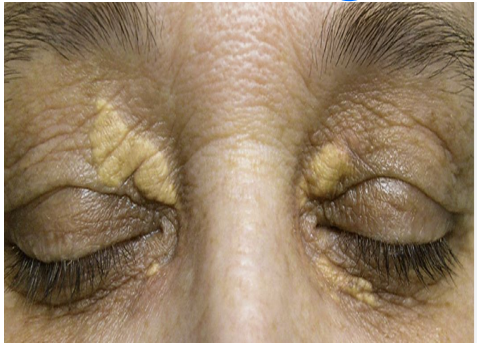
c. Type III

d. Type V

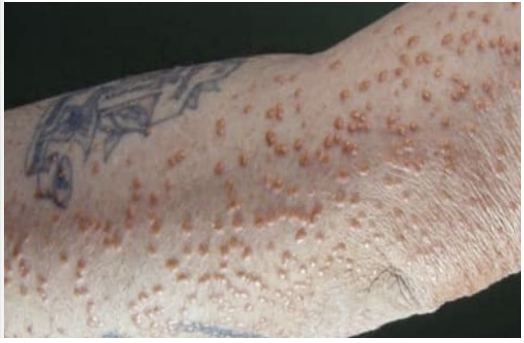


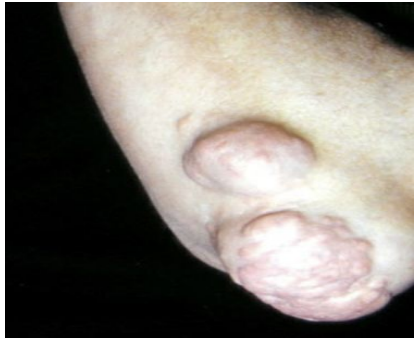
Xanthelasma	Eyelids	Type II / Normal
Plane	Flat patches	Type III / FH
Palmar	Palmar creases	Type III
Tuberous	Elbows/ knees	Type III
Tendon	Achillles	Type IIa
Eruptive	Buttocks	TG↑ (I,IV,V)

xanthelcma : (n)



Eruptive : I





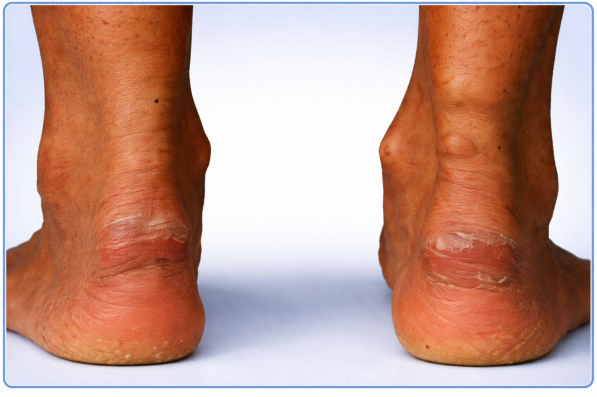
20. The following xanthomas are seen in which of the following hyperlipoproteinemia

a. Type I

b. Type II

c. Type III

d. Type V



21. Which of the following will convert 25-OH D3 to 1,25-OH_2 D3?

1α Hydroxylase

a. UV light

b. Sunlight

c. PTH

d. Calcitonin

P.K.U

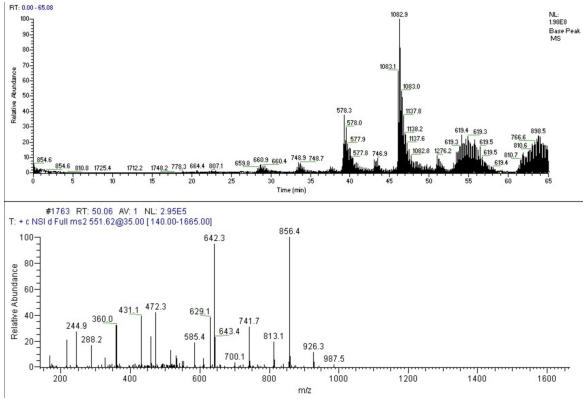
22. Neonate develops intractable vomiting on day 15 of life with recurrent episodes of seizures. Examination shows musty Odor from skin blond hair and blue iris. Which of the following is in investigation of choice for this condition?

a. Guthrie test

b. Ferric chloride test

c. Tandem mass spectrophotometry

d. Dried heel pad blood



23. Substrate level phosphorylation is done which class enzyme?

a. Carboxylase CO_2 + (Biotin)

b. Kinase

c. Dehydrogenase Oxidation - Reduction

d. Hydroxylase add OH^-

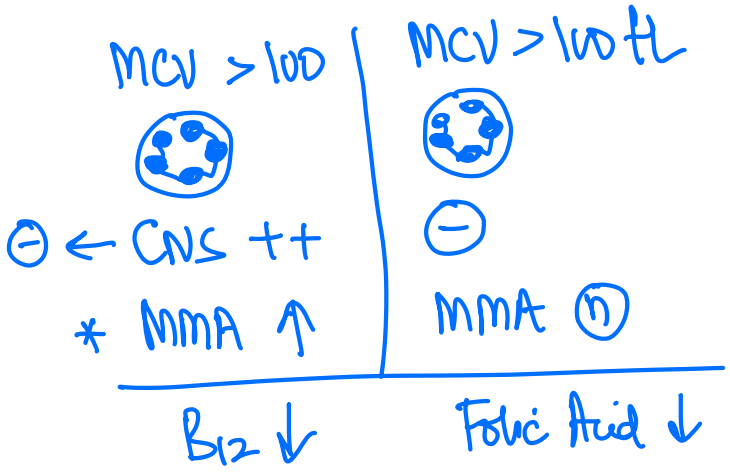
24. Patient is having macrocytic megaloblastic anaemia with hypersegmented neutrophils on microscopic examination of peripheral smear. CNS examination is normal. Work up shows normal methylmalonic acid level. This is seen in?

a. Folic acid deficiency

b. Vitamin B12 deficiency

c. Copper deficiency

d. Iodine deficiency



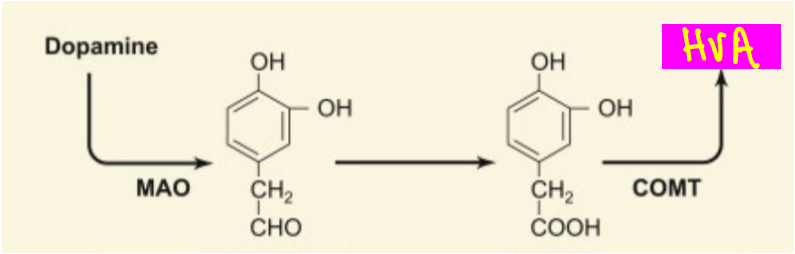
25. Major final end product of catecholamine dopamine is which of the following?

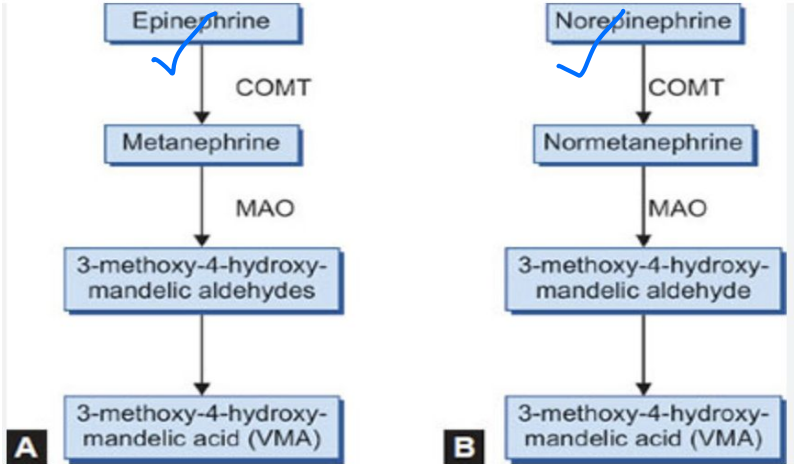
a. Vanillylmandelic acid

b. Homovanillic acid

c. Metanephrine

d. Di-hydroxy-phenylacetic acid





26. A 4-year-old boy child is diagnosed with haemolytic anaemia. Peripheral Smear shows RBC inclusion in supravital staining. Which enzyme deficiency is likely?

a. Glucose 6 phosphatase

b. Glucose 6 phosphate dehydrogenase

c. Pyruvate decarboxylase

d. Phosphophenol pyruvate carboxykinase

HEINZ Bodies = denatured Hb

COLLAGEN #

27. Child is noted to have corkscrew hair and perifollicular haemorrhages. This is seen in which of the following:

a. Kwashiorkor

b. Vitamin K deficiency

c. Vitamin C deficiency

d. Porphyria

plasma ascorbate levels ↓

vit C: scurvy

1. Perifollicular bleeding
2. Bone pain: frog leg app: pseudo paralysis
3. gum bleeding

XRay knee: metaphyseal ++

DVT

28. Patient presents with diarrhoea, depression and diabetes mellitus. On examination erosive red-brown plaques are noticed around lips and lower abdomen. This is common presentation seen in

a. Farmer who is corn eater X

b. Patient with PNET

GLUCAGONOMA: Necrolytic migratory erythema (dermatitis)

c. Athlete who is vegan

d. Bodybuilder consuming egg whites

29. Which Vitamin deficiency leads to loss of proprioception and vibration sense?

a. Vitamin A

b. Vitamin D

c. Vitamin E

d. Vitamin K

Retinal #
Bone pain
myelin formation # , RBC membrane #
ATAXIA , ACANTHOCYTES

30. Thiamine pyrophosphate is a cofactor for all of the following except?

a. Pyruvate dehydrogenase

b. Alpha ketoglutarate

c. Branched chain keto acid dehydrogenase

d. Trans aldolase

TRANS-KETOLASE

31. RBC glutathione reductase levels are used to asses deficiency of which of the following vitamin?

a. B2
b. B3
c. B9
d. B12

FIGLU ↑
MMA ↑

RBC Transketolase = B_1
// glutathione Reductase = B_2
// amino aspartate = B_6

Histidine metabolism

32. Form-imino-glutamic acid levels in urine is used for assessment of which of the following?

a. Vitamin A

b. Vitamin H

c. Vitamin B9

d. Vitamin B12

33. Which of the following is not a component of creatinine?

a. Glycine

b. Arginine

c. Methionine

d. Glutamic acid

```
Glycine + Arginine
        ↓  (kidney – transamidination)
Guanidinoacetate + Ornithine
        ↓  (liver – methylation by SAM from Methionine)
Creatine
        ↓  (muscle – phosphorylation)
Phosphocreatine
        ↓  (non-enzymatic)
Creatinine (excreted in urine)
```

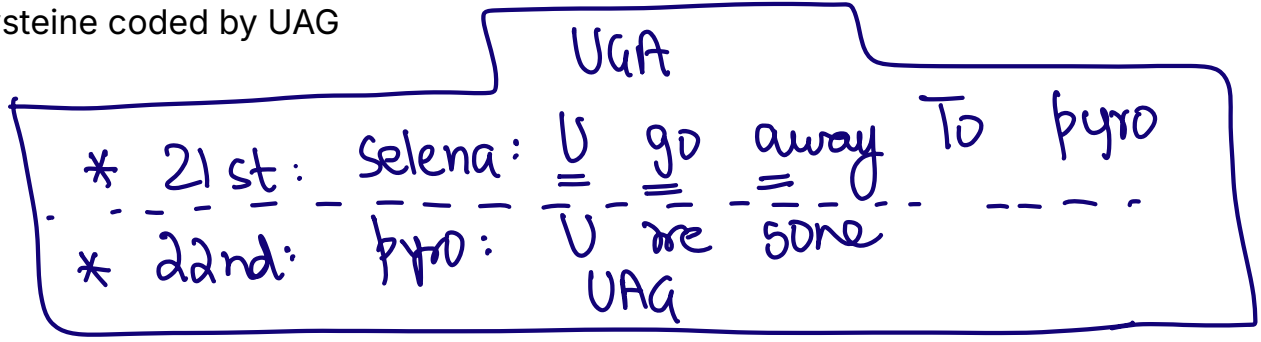
34. Which is correct about 22nd amino acid?

a. Pyrrlolysine coded by UAG

b. Pyrrolysine coded by UGA

c. Selenocysteine coded by UGA

d. Selenocysteine coded by UAG



35. Purely ketogenic amino acid is which of the following?

a. Leucine

b. Lysine

c. Aspartic acid

d. Histidine

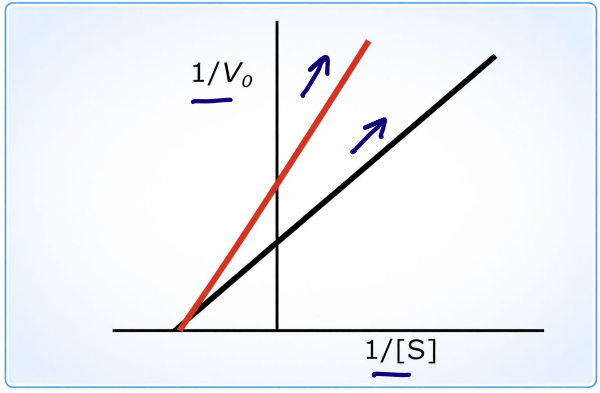
36. The red line in Lineweaver burk plot indicates which of the following

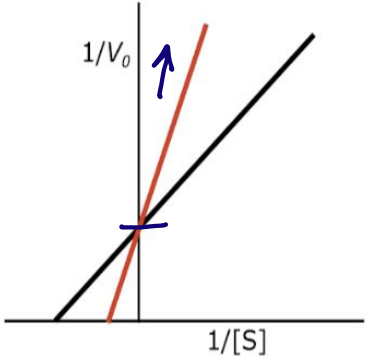
a. Competitive inhibition

b. Un-competitive inhibition

c. Non competitive inhibition

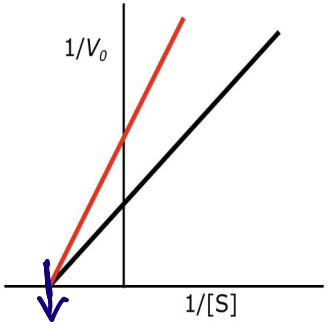
d. Feedback inhibition





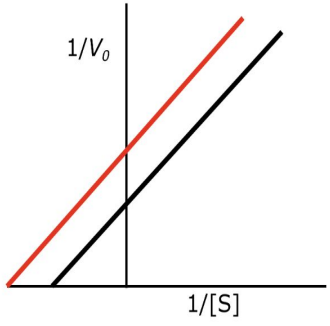
* C: y axis, slope ↑, km ↑

* NC: x axis, slope same



Noncompetitive Inhibition

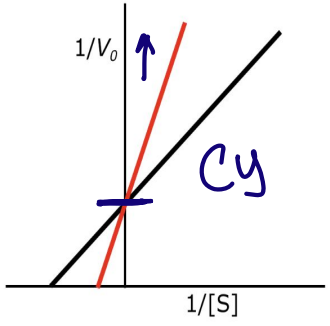
Decreased V_{max}= increased $1/V_{max}$



Uncompetitive Inhibition

Decrease K_m = increased $1/K_m$

Decreased V_{max}= increased $1/V_{max}$



Competitive Inhibition

Increase K_m = decreased $1/K_m$

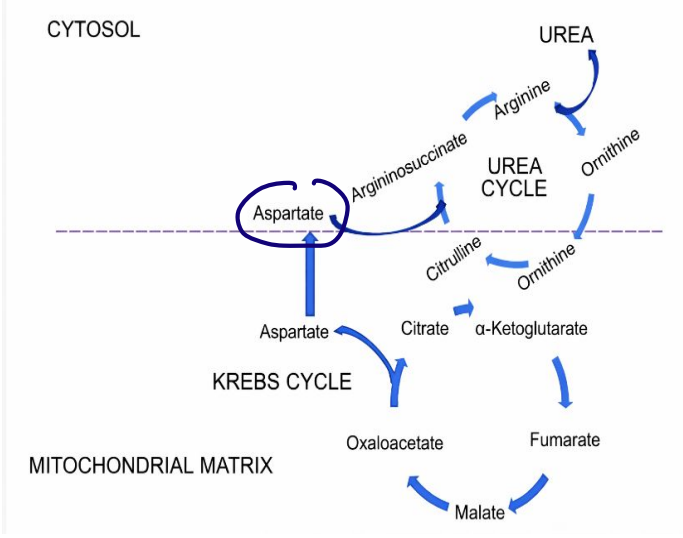
37. Urea bicycle is linked to TCA cycle via

a. Fumarate and aspartate

b. Fumarate and lactate

c. Fumarate and pyruvate

d. Fumarate with arginase



38. Oxidative deamination producing ammonia occurs in which organ?

a. Kidney

b. Liver

c. Skeletal muscle

d. Brain

39. The most common urea cycle defect is which of the following?

a. Carbamoyl phosphate synthetase-1

b. Ornithine transcarbamoylase

c. Arginase

d. Arginosuccinate

40. Patient presents with recurrent kidney stones. Urine microscopy shows hexagonal crystals. Which is the biochemical investigation to determine the type of kidney stone?

a. NCCT KUB

b. Urine pH

c. Urine aminoaciduria screen

d. Urine Cyanide nitroprusside test

CYSTINE STONES

 * Sulphur containing AA

41. Which is correct dietary intervention for PKU?

a. High tyrosine diet

b. Low tyrosine diet

c. Complete elimination of phenylalanine in diet

d. Add phenylalanine supplements in diet

42. Which is correct diagnosis of Tall marfanoid habitus with kyphosis, hypopigmented -skin and high urinary cyanide nitroprusside test?

a. Alkaptonuria

b. Tyrosinemia

c. Homocystinuria

d. PKU

* INFERO - NASAL : ectopic lentis

43. A newborn with lethargy and sweet-smelling urine has defect in:

a. Branched-chain α-ketoacid dehydrogenase

b. Hom gentisate oxidase

c. Phenylalanine hydroxylase

d. Tyrosinase

44. A tall, thin child with infero-nasal lens dislocation and thrombosis likely lacks which of the following enzymes

HOMOCYSTINEMIA ↑CAD

a. Cystathionine β-synthase

b. Methionine synthase

c. Tyrosinase

d. Galactose-1-P uridyltransferase

45. A neonate with jaundice, vomiting and E. coli sepsis lacks:

a. Galactose-1-phosphate uridyltransferase

b. Galactokinase

c. Aldose reductase

d. UDP-galactose epimerase

Rx :- Soya-Milk

~~lactose~~

46. A child with coarse facies, hepatosplenomegaly and corneal clouding lacks:

a. α-L-iduronidase

b. Iduronate sulfatase

c. Galactosidase

d. Ceramidase

HURLER

IDURASE

47. A 4-year-old boy presents with progressive developmental delay, hepatosplenomegaly, coarse facial features, and frequent ear infections. His corneas are clear and parents report increasing aggressive behaviour. Deficiency of which enzyme is most likely?

a. α-L-Iduronidase

b. Iduronate sulfatase

c. Arylsulfatase A

d. β-Galactosidase

48. A nucleotide differs from a nucleoside by presence of which additional component?

a. Nitrogen base

b. Sugar

c. Phosphate group

d. Ribose

49. DNA replication in humans is described as semi-conservative because each new DNA contains:

a. Two old strands

b. Two new strands

c. One old and one new strand

d. Randomly mixed strands

50. The enzyme that unwinds the DNA double helix ahead of the replication fork is:

a. Ligase

b. Helicase

c. Primase

d. Telomerase

51. A researcher needs to synthesize a short RNA primer to start DNA replication. Which enzyme helps?

a. Primase

b. Ligase

c. Topoisomerase

d. Endonuclease

52. During PCR, the step in which DNA strands are separated by heating is known as:

a. Annealing

b. Extension

c. Denaturation

d. Amplification

53. During PCR, primers bind to the single-stranded DNA template during which step?

a. Extension

b. Annealing

c. Denaturation

d. Cooling

54. The enzyme responsible for adding nucleotides during mRNA synthesis is:

a. DNA polymerase

b. RNA polymerase

c. Reverse transcriptase

d. Ligase

55. What is correct about composition of glycosaminoglycans?

a. Uronic acid plus amino sugars

GAG

b. Uronic acid and hyaluronic acid

c. Uronic acid and galactose

d. Uronic acid and glucose

56. Corneal transparency is maintained by which of the following

a. Type IV collagen

b. Type III collagen

c. Keratan sulfate

d. Dermatan sulfate

57. Which is the most abundant GAG?

a. Chondroitin

Cartilage

b. Keratan sulphate

c. Dermatan sulphate

d. Heparan sulfate

58. Which of the following will inhibit GLUT4?

a. Empagliflozin SGLT2i

b. Phloretin

c. Phlorizin

d. Inulin

* P-TIN blocks passive transport ie GLUT
ZIN blocks Zymport ie SGLT

59. A 50-year-old patient is detected to have familial renal glycosuria. The defect is present at which of the following sites?

a. Duodenum

b. Pancreas

c. PCT

d. DCT

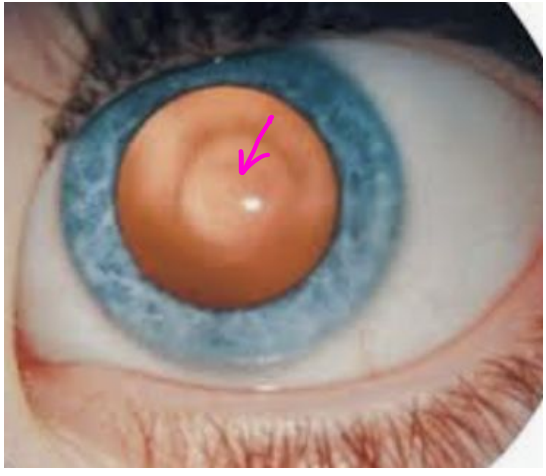
60. Oil droplet cataract in galactosemia is due to accumulation of which of the following products *

a. Galactokinase

b. Galactose 1-uridyl-transferase

c. Galactitol

d. UDP-galactose



61. A 1 month old child has vomiting episodes every time fruit juice or honey is given to him. Fructose restriction was advised and child showed weight gain. Which enzyme is deficient?

a. Aldolase A

b. Aldolase B

HFI

c. Fructokinase

d. Triose kinase

62. PUFA is least in which of the following cooking oils

a. Safflower oil

b. Sunflower oil

c. Coconut oil

d. Rapeseed oil

63. Most dense lipoprotein with least TG content is

a. Chylomicrons

b. LDL

c. HDL

d. VLDL

Healthy Dense Lipoproteins" HDL = most **H**eavy/**D**ense, **L**east TG

64. Function of hormone sensitive lipase in adipocytes?

a. Lipolysis

b. Lipogenesis

c. Lipid conjugation

d. Lipid deconjugation

65. Which is correct about Warburg effect? CANCER cells : MITOTIC RATE ++

a. Aerobic glycolysis

b. Anaerobic glycolysis

c. Aerobic gluconeogenesis

d. Anaerobic gluconeogenesis

Pure waste of resources

→ Produces faster ATP though less

→ Lactate favour cancer cell survival and kills normal cells

→ Promotes angiogenesis

66. Phosphofructokinase 1 is rate limiting step in _____

a. Gluconeogenesis

b. Glycogenolysis

c. Embden meyerhof pathway

d. Cori cycle

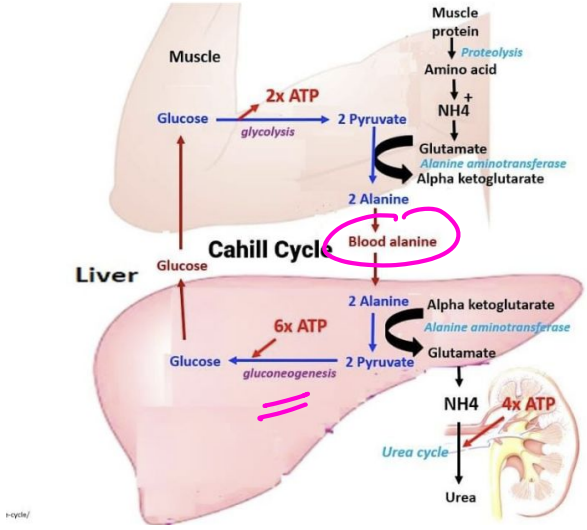
67. Which amino acid plays dominant role in Cahill cycle

a. Glycine

b. Alanine

c. Proline

d. Histidine



68. Which of the following is an uncoupling agent in electron transport chain?

a. Alcohol

b. Rotenone

c. Antimycin

d. Hydrogen sulfide

mitochondria : cell membrane #

69. All of the following processes occur in both mitochondria and cytoplasm except?

a. Heme synthesis

b. Urea synthesis

c. Gluconeogenesis

d. TCA cycle

Quenching = rapid cooling or **chemical stabilization** to **prevent re-annealing** and keep DNA single stranded.

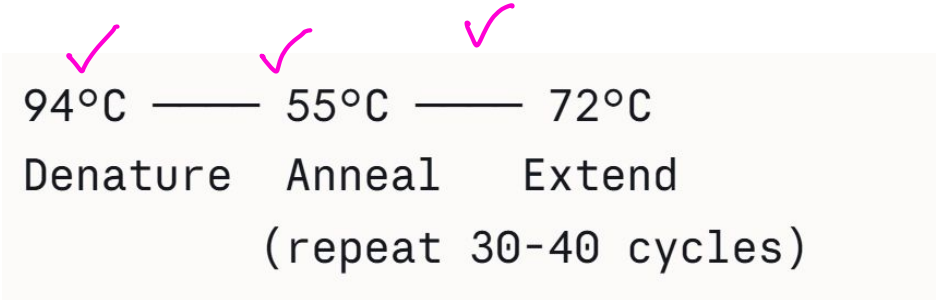
70. _____ causes quenching of denatured DNA?

a. Slow cooling

b. Rapid cooling

c. Slow heating

d. Rapid heating



71. DNA helicase is defective in:

a. Bloom syndrome

b. Ataxia telangiectasia

c. Friedreich ataxia

d. Li Fraumeni syndrome

72. Which type of PCR quantifies DNA amplification in real time using fluorescent dyes?

a. Nested PCR

b. RT-PCR

c. qPCR

d. Multiplex PCR

73. PCR type that uses multiple primer sets to amplify different genes simultaneously is:

a. Nested PCR

b. Multiplex PCR

c. qPCR

d. Digital PCR

74. A highly specific PCR using two successive rounds of amplification to reduce false positives is:

a. Nested PCR

b. qPCR

c. Multiplex PCR

d. Hot-start PCR

Nested	✓	↑ Specificity — two successive rounds
qPCR (Real-time)	*	Quantification of DNA/RNA
Multiplex	✓	Detect multiple targets simultaneously
Hot-start		Reduce non-specific amplification at start
RT-PCR		RNA → cDNA → amplify (detect mRNA)
ARMS PCR		Detect point mutations
Digital PCR		Absolute quantification

75. DNA matching used for identification and comparison among family members is based on analysis of which genetic marker?

a. Short tandem repeat (STRs)

b. Single nucleotide polymorphisms (SNPs)

c. Ribosomal RNA genes

d. Mitochondrial DNA

THANK YOU