

Clinical Biochemistry – NEET PG Q&A;

1. A chronic alcoholic presents with confusion, ataxia, and ophthalmoplegia. Which vitamin deficiency is most likely?

Answer: Vitamin B1 (Thiamine)

Explanation: Wernicke's encephalopathy is due to thiamine deficiency → common in chronic alcoholism. Triad: confusion, ataxia, ophthalmoplegia.

2. A patient with megaloblastic anemia and neurological symptoms is most likely deficient in:

Answer: Vitamin B12

Explanation: Both folate and B12 deficiency cause megaloblastic anemia, but neurological features (subacute combined degeneration) are unique to B12 deficiency.

3. A 10-year-old child presents with mental retardation and mousy odor in urine. Most likely diagnosis is:

Answer: Phenylketonuria (PKU)

Explanation: Deficiency of phenylalanine hydroxylase → accumulation of phenylalanine → mental retardation + mousy odor urine.

4. A patient's urine turns black on standing. Which enzyme deficiency is likely?

Answer: Homogentisate oxidase

Explanation: Alkaptonuria due to deficiency of homogentisate oxidase → homogentisic acid excretion → black urine on standing.

5. A newborn presents with failure to thrive, jaundice, and cataracts. Reducing sugars are present in urine. The likely cause is deficiency of:

Answer: Galactose-1-phosphate uridyl transferase

Explanation: Classical galactosemia due to deficiency of GALT enzyme → hepatomegaly, jaundice, cataracts, mental retardation.

6. A patient presents with hyperuricemia and self-mutilation behavior. Most probable diagnosis is:

Answer: Lesch–Nyhan syndrome

Explanation: X-linked recessive disorder due to HGPRT deficiency → ↑ uric acid, neurobehavioral problems (self-mutilation, aggression).

7. A person presents with recurrent infections, chronic diarrhea, and failure to thrive. Adenosine deaminase deficiency is found. This condition is:

Answer: Severe combined immunodeficiency (SCID)

Explanation: ADA deficiency leads to accumulation of toxic metabolites in lymphocytes → defective immunity → SCID.