QUIZ

IEM

1

- **Sepsis with which pathogen suspects IEM**

a) E.Coli.

b) Staph aurieus.

c) Streptpcocci.

d) Pnuemococci

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An IEM in which liver damage and in chronic cases liver cell carcinoma results from a deficiency of fumaryl acetoacetate hydrolase.

a) Ornithinuria.
b) Oxalic aciduria.
c) Tyrosinaemia type 1.
d) Homocysteinuria type1.

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- **Alopecia/ Dermatitis is seen in**

a) Arginosuccinic aciduria

b) Citrullinemia

c) Hawkinsuria

d) Cysteinosis

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- **Agenesis of corpus callosum/ cortical cysts are seen in**

a) Pyruvate kinase deficiency.

b) ketoacid decarboxylase.

c) PDH deficiency.

d) α-keto glutarate dehydrogenase

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- **Glutaric aciduria Type I shows**

a) Macrocephaly

b) Microcephaly

c) Anencephaly.

d) Elephant face

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- **Sweaty feet odour is diagnostic of**
- Isovaleric aciduria

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- **Which of the following is employed as therapeutic measure in treatment of IEM**

a) Dialysis

b) Exchange trasfusion

c) Plasmapheresis

d) All of the above

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Of the following, the MOST appropriate statement to include in your discussion of PKU screening is that:

a) Affected infants cannot be breastfed.

b) Dietary therapy should not be initiated until screening results are confirmed.

c) It relies on identification of a specific mutation in the phenylalanine hydroxylase gene.

d) To be most reliable, the specimen should be obtained after 24 hours of age.

9.

- **Renal cysts are seen in**

a) Cysteinuria

b) Alkaptonuria

c) Glutaric aciduria.

d) Malonic aciduria

10

- **Lactic Acidosis is seen glycogen storage disorder type**

a) Type 1.

b) Type 2.

c) Type 3.

d) Type 4

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- **Lactate to pyruvate ratios are**

a) 10:1 to 20:1.

b) 1:1 to 2:1.

c) 0.5:1 to 10:2.

d) 1:1.5 to 2.5:1

- **OTC Deficiency**

a) Xlinked dominant

b) Xlinked trait

c) Autosomal trait

d) Autosomal dominant

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- **Tetrahydrobiopterin is required for the hydroxylation of**

a) Phenylalanine
b) Proline
c) Lysine
d) All of the above

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Which of following is/ are correctly linked with their deficiency disease

a) Glucose-6- phosphatase - lactic alkalosis

b) Muscle phosphorylase- fasting hypoglyceamia

c) Galc -1- P uridyltransferase – cataract

d) Fructokinase- heriditory fructose intolerance

15

- **NH_3 scavengers used to treat urea cycle disorders are**

a) Phenyl alanine.

b) Phenyl lactate.

c) Phenyl butyrate.

d) Phenyl acetate.

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- **Smell of rotting fish is diagnostic of which Aminoaciduria**
- Trimethylaminuria

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- **Supplimentation with what prevents protein catabolism**

a) High protein diet

b) Low calorie diet

c) High calorie diet

d) Multivitamin high dose diet.

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- Albinism an inborn error of metabolism is due to lack of

  a) Tyrosine carboxylase
  b) Homogenestic acid oxidase
  c) Phenyl alanine hydroxylase
  d) Tyrosinase

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- **Maple syrup urine disorder is an inborn error of metabolism of**

a) Sulphur containing amino acids

b) Aromatic amino acids

c) Branched chain amino acids

b) Dicarboxylic amino acids

20

Phenyl pyruvic acid in urine is detected by

a)Ferric chloride test
b) Guthries test
c) Gerharts test
d) VMA urine

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- **Presence of which sugar makes Soy the recommended supplement in patients with PKU**

a) Glucose

b) Sucrose

c) lactose.

d) Maltose.

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Enzyme deficient in homocystinuria

a) Cystathionine hydroxylase
b) Cystathionine synthetase
c) Cystathionine reductase
d) Cystathionine isomerase

23

- **Shaken baby like syndrome (IC bleeding, Retinal bleeding) is seen in which IEM**

a) Methylmalonyl Aciduria

b) Glutaryl aciduria

c) Isovaleric aciduria

d) Tyrosinemia

24. A 9 year old male is brought to the emergency room due to vomiting and lethargy shortly after a birthday party. He had had several bouts of vomiting in the past, usually after consuming candy or soft drinks. Labs reveal elevated AST and ALT. What is the most likely diagnosis?

a) Hereditary fructose intolerance

b) Glycogen storage disease Type II

c) Fatty acid oxidation defect

d) Fabry disease

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Aminoacid carrier defect is found in

a) MSUD
b) Alkaptonuria
c) Cystinuria
d) Homocystinuria

26

- **Smell of Cabbage is diagnostic of malabsorption of which amino acid**
- Methionine

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- **Nonketotic hyperglycinemia is seen in**

a) Glycine cleavage system
b) Deficiency of Lipoamide dehydrogenase
c) Glycine decarboxylase deficiency
d) Methyl transferase deficiency.

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- **Urinary excretion of homogentisic acid is increased in**

a) Tyrosinaemia
b) Alkaptonuria
c) Phenyl ketonuria
d) Homocystinuria

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- **Musty smell is diagnostic of which aminoaciduria**
- PKU

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- **Hypoketotic hypoglycemia is seen in**

a) LCFA & MCFA acyl CoA dehydrogenase deficiency.

b) LCFA & MCFA acyl CoA reductase deficiency.

c) LCFA & MCFA acyl CoA decarboxylase deficiency.

d) LCFA & MCFA transacylase deficiency.

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- **In galactosemia , there is deficiency of**

a) Galactosidase
b) Galactose 1-phosphate uridyl transferase
c) Aldolase reductase
d) UDP-Hexose 4 epimerase

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- **Refsum’s disease is due to accumulation of**

  a) Galactose

  b) Sorbitol

  c) Phytanic acid

  d) Sphingomyelinase