قائمة الاسئلة

الاختلالات الجينية في التمثيل الغذائي - ()- المستوى الثالث -قسم التغذية العلاجية والحميات - - كلية الطب والعلوم الصحية - الفترة الخامسة- درجة ا

د/ نوال الحنحنة

1) What is the primary benefit of antioxidants in the diet?
   1) - Raise cholesterol levels
   2) - Prevent muscle gain
   3) + Protect cells from damage
   4) - Increase blood pressure
2) Coenzyme Q and creatine supplementation are crucial in management of
   1) + Mitochondrial disease
   2) - Lipid metabolism disorder
   3) - Niemann-Pick disease
   4) - Wilson disease
3) PKU is caused by a defect in the gene of synthesis hepatic enzyme
   1) - Branched chain alpha ketoacid (BKA) dehydrogenase complex
   2) - Homogentisic dioxygenase
   3) + Phenylalanine hydroxylase
   4) - Fumarylacetoacetase
4) Which of the following is NOT a common symptom of hemochromatosis?
   1) - Joint pain
   2) - Diabetes
   3) + Anemia
   4) - Chronic fatigue
5) What is the main dietary treatment for individuals diagnosed with celiac disease?
   1) - Low-carbohydrate diet
   2) + Gluten-free diet
   3) - High-protein diet
   4) - Lactose-free diet
6) Chylomicronemia syndrome characterized by
   1) - Hypoglycemia
   2) + Massive hypertriglyceredemia
   3) - Hypercholesterolemia
   4) - Metachromatic leukodystrophy
7) The patients present with acidosis, vomiting, convulsions and coma.
   1) - Carnitine Transporter Deficiency (CTD)
   2) - Medium-chain Acyl-CoA Dehydrogenase Deficiency.
   3) + Organic Acidurias
   4) - All of the above
8) Example of mitochondrial diseases
   1) + Leber’s hereditary optic neuropathy (LHON)
   2) - Gaucher disease
   3) - Tay-Sachs disease
   4) - Fabry disease
9) OTC (Ornithine Transcarbamylase) Deficiency is one of
   1) - Mitochondrial diseases
   2) - Lipid metabolism disorders
   3) + Urea cycle disorders
   4) - Carbohydrate metabolism disorders

10) Increase in blood of this class of lipoproteins is beneficial to ward off coronary heart disease:
1) - HDL
2) + LDL
3) - IDL
4) - VLDL

11) Mitochondria diseases are worse when the defective present in the
1) - Cells of muscles
2) - Cerebrum
3) - Nerves
4) + All of the above

12) Maple Syrup Urine Disease is caused by a defect in the gene of synthesis enzyme
1) + Branched chain alpha ketoacid (BKA) dehydrogenase complex
2) - Homogentisic dioxygenase
3) - Phenylalanine hydroxylase
4) - Fumarylacetoacetase

13) Which of the following symptoms is NOT typically associated with celiac disease?
1) - Chronic diarrhea
2) + Weight gain
3) - Abdominal pain
4) - Fatigue

14) The cholesterol serves as the precursor for the following biosynthetic pathways, EXCEPT
1) - Bile acid synthesis
2) - Steroid hormone synthesis
3) - Aldosterone synthesis
4) + Thyroid hormone synthesis

15) In nutritional management of hemochromatosis, which of the following foods is generally safe and recommended?
1) - Red meat
2) - Spinach
3) + Dairy products
4) - Citrus fruits

16) Lactase deficiency
1) - It is a deficiency of lactase enzyme
2) - Congenital
3) - Acquired
4) + All of the above

17) Zinc supplements may also be prescribed for those with
1) - Gout disease
2) + Wilson disease
3) - Celiac disease
4) - Hemolytic disease

18) It is a congenital condition in which glucose and galactose are absorbed slowly
1) - Disorder in phospholipid metabolism
2) + Monosaccharide malabsorption
3) - Cholesterol metabolism disorder
4) - Fatty acid metabolism disorder

19) Which of the following minerals is often used in the treatment of Wilson's disease to inhibit copper absorption?
1) - Magnesium

2) - Iron
3) + Zinc
4) - Calcium

20) Tyrosinaemia type 1 is caused by a defect in the gene of synthesis enzyme
1) - Branched chain alpha ketoacid (BKA) dehydrogenase complex
2) - Homogentisic dioxygenase
3) - Phenylalanine hydroxylase
4) + Fumaryl acetoacetase

21) Which nutrient deficiency is common in untreated celiac disease due to malabsorption?
1) - Vitamin C
2) - Calcium
3) + Iron
4) - Zinc

22) Sphingolipidoses is the disorder of?
1) + phospholipid metabolism
2) - ketone body metabolism
3) - cholesterol metabolism
4) - fatty acid metabolism

23) Treatment of hyperammonemia
1) - Decrease protein intake (a source of ammonia)
2) - Intravenous sodium phenylacetate and sodium benzoate
3) - Acidification of the intestinal lumen using lactulose
4) + All of the above

24) Inborn errors of metabolism are referred to as
1) - Congenital metabolic diseases
2) - Inherited metabolic diseases
3) + All of the above
4) - None of above

25) Which type of nucleic acid is primarily affected in diseases like Lesch-Nyhan syndrome and gout
1) - DNA only
2) - RNA only
3) + Both DNA and RNA
4) - Mitochondrial DNA

26) Kayser-Fleischer Rings is of the more unusual presentations of
1) - Gaucher disease
2) - Tay-Sachs disease
3) - Niemann-Pick disease
4) + Wilson disease

27) Favism
1) - Congenital sex-linked inducible hemolytic disease
2) - Inherited deficiency of glucose-6-phosphate dehydrogenase
3) - Deficiency of NADPH.H+
4) + All of the above

28) Which genetic testing result confirms a diagnosis of hereditary hemochromatosis?
1) - Mutation in the CFTR gene
2) + Mutation in the HFE gene, specifically C282Y or H63D
3) - Absence of the HFE gene
4) - Extra copy of the HFE gene

29) Which of the following is the primary cause of celiac disease?

1) + Gluten consumption
2) - Bacterial infection
3) - Genetic mutation unrelated to diet
4) - Vitamin D deficiency

30) The disorder of copper metabolism is
1) - Mutations to transporter (ATP7B gene) within hepatocytes
2) - Wilson disease
3) - An autosomal recessive disorder
4) + All of the above

31) Which is caused by the combined deficiency of hexosaminidase A and B.
1) - Tay-Sachs disease
2) - Niemann-Pick disease
3) - Gaucher disease
4) + Sandhoff's disease

32) Giving a diet low in branched chain amino acids is management in
1) - Alkaptonuria
2) - Cystinuria
3) + Maple syrup urine disease
4) - Parkinson's Disease

33) Gaucher's disease cause progressive brain damage and seizures.
1) - Type I
2) - Type II
3) + Type III
4) - None of the above

34) A disease that result in loss of hair and skin pigments is
1) - Alkaptonuria
2) - Cystinuria
3) + Albinism
4) - Parkinson's Disease

35) What dietary recommendation is often advised for patients with gout, a disorder related to purine metabolism?
1) - High-protein diet
2) + Low-purine diet
3) - High-calcium diet
4) - Low-zinc diet

36) Which dietary restriction is recommended for managing Wilson's disease?
1) - High-protein diet
2) + Low-copper diet
3) - High-sodium diet
4) - Low-calcium diet

37) Autoimmune disorder that results in damage to the lining of the small intestine
1) + Celiac disease
2) - Maple syrup urine disease
3) - Parkinson's Disease
4) - Gaucher disease

38) Which lifestyle modification is recommended for a patient with hemochromatosis to prevent iron overload?
1) - Consuming high-vitamin C supplements
2) + Drinking tea or coffee with meals
3) - Avoiding all forms of dairy

4) - Consuming more red meat

39) Which of the following is the main enzyme deficiency in Lesch-Nyhan syndrome, a nucleic acid metabolism disorder?
1) - Adenosine deaminase
2) + Hypoxanthine-guanine phosphoribosyltransferase (HGPRT)
3) - Xanthine oxidase
4) - Glucose-6-phosphate dehydrogenase

40) Which test is commonly used to diagnose Wilson's disease?
1) - Serum copper level
2) - Blood glucose test
3) + Urinary copper excretion test
4) - Serum zinc level

41) What is the primary cause of Wilson's disease?
1) - Excessive dietary copper intake
2) + Genetic mutation in the ATP7B gene
3) - Deficiency of ceruloplasmin
4) - High zinc intake

42) Bronze Colored Skin is seen in
1) + Hereditary hemochromatosis
2) - Albinism
3) - Wilson disease
4) - Celiac disease

43) Duodenal mucosa histology test is to diagnosis of
1) - Lysosomal storage diseases
2) - Maple syrup urine disease
3) - Parkinson's Disease
4) + Celiac disease

44) What is the primary cause of hereditary hemochromatosis?
1) - Excessive dietary iron intake
2) + Genetic mutation in the HFE gene
3) - Chronic liver disease
4) - High vitamin C intake

45) In which condition is xanthine oxidase activity critical for disease management?
1) - Lesch-Nyhan syndrome
2) + Gout
3) - Phenylketonuria
4) - Cystic fibrosis

46) Which of the following organs is most commonly affected by iron overload in hemochromatosis?
1) - Pancreas
2) - Kidneys
3) + Liver
4) - Spleen

47) Which of the following is a common clinical sign of Wilson's disease?
1) - High blood pressure
2) - Joint pain
3) + Kayser-Fleischer rings
4) - Skin rash

48) Which type of diet modification is beneficial in managing the symptoms of Adenosine deaminase deficiency?

1) - Low-fat diet
2) + High-antioxidant diet
3) - High-protein diet
4) - Low-glucose diet

49) Which lifestyle modification is most recommended to support mental health?
1) - Increased screen time
2) + Physical activity
3) - Eating sugary snacks
4) - Irregular sleep patterns

50) Which vitamin is crucial in helping the body absorb calcium, especially for bone health?
1) - Vitamin C
2) - Vitamin B12
3) + Vitamin D
4) - Vitamin K