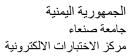




قائمة الاسئلة

## الاختلالات الجينية في التمثيل الغذائي - ()- المستوى الثالث -قسم التغذية العلاجية والحميات - للية الطب والعلوم الصحية - الفترة الخامسة-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





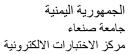
- 10) Increase in blood of this class of lipoproteins is beneficial to ward off coronary heart disease:
  - 1) HDL
  - 2) + LDL
  - 3) IDL
  - 4) VLDL
- 11) Mitochondria diseases are worse when the defective present in the
  - 1) Cells of muscles
  - 2) Cerebrum
  - 3) Nerves
  - 4) + All of the above
- 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
- 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
- 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
- Which of the following minerals is often used in the treatment of Wilson's disease to inhibit copper absorption?
  - 1) Magnesium

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- 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?

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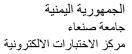
- 1) + Gluten consumption
- 2) Bacterial infection
- 3) Genetic mutation unrelated to diet
- 4) Vitamin D deficiency
- 30) The disorder of copper metabolism is
  - 1) Mutations to transporter (ATP7B gene) within hepatocytes
  - 2) Wilson disease
  - 3) An autosomal recessive disorder
  - 4) + All of 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
- A disease that result in loss of hair and skin pigments is
  - 1) Alkaptonuria
  - 2) Cystinuria
  - 3) + Albinism
  - 4) Parkinson's Disease
- 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
- 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
  - Gaucher disease
- 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

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- 4) Consuming more red meat
- 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
- 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
- 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
- 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
- Which type of diet modification is beneficial in managing the symptoms of Adenosine deaminase deficiency?

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