بسم الله الرحمن الرحيم

541 BCH

نموذج من الإمتحان النهائي لمادة حيود الأيض

Please mark the correct statements with (T) and the incorrect one with (X):

1- Hypoammonemia caused by the defect of all urea cycle enzymes.
2-The diploid organism contains two copies of every gene, excepting those carried on the sex chromosomes.
3- G6PD deficiency is a sex-linked affecting males and being carried by females
4- In diabetic ketosis, liver produces large amounts of ketone bodies, which lead to decrease in pH then impairs tissue function such as central nervous system.
5- For an autosomal recessive trait to be expressed, both copies of a gene pair must be present in a mutant form.
6- Homogentisate is abnormal intermediate in the degradation of phenylalanine and tyrosine amino acids.
7- A person who is homozygous for aa has inherited the a allele from his great-grandmother then transmitted through one parent.
8- When 2,4-Dinitrophenylhydrazone react with α-KA the result will be 2,4-dinitrophenylhydrozone derivative.
9- The component activity in replication that reduces the number of mis- incorporated bases known as postreplication repair system.
10-Phenylketonuria symptoms including the decrease in brain weight, myelination of their nerves is defective and their reflexes are hyperactive.
11-When the transferase is not active in the lens of the eye then the presence of aldose Reductase causes the accumulating galactose to be reduced to galactitol.
12-Porphyrin biosynthesis is taking place in liver and phagocytes and either site might be the disorder site.
13-The homologue is referred to each chromosome is derived from a different parent which is known as homozygous.
14-Tyrosinemia symptoms are absence of pigmentation, scoliosis and seizures.
15-Type 2 DM is a homogeneous syndrome resulting from defects of both insulin secretion and action.
16-hypolipidemia phenotypes result from defects in either lipoprotein synthesis, metabolism, or uptake by tissues.
17-The possible cause of hyperammonemia is that elevated levels of glutamate formed from NH4+ and glutamine.
18-Recessive mutation is known in which both alleles must be mutant.
19-Carnitine deficiency can affect the cardiac muscle and result in poor cardiac contraction known as cardiomyopathy.
20- The congenital erythropoietic porphyria causes prematurely erythrocytes

hemolysis which caused by insufficient cosynthase.

21-A severe mutation might occur during recombination events when some pieces of DNA can move to different positions in the genome.

22-In MODY2, the hyperglycemia linked with GCK mutation is often mild.

23-The cystathionine-β-synthase deficiency caused homocystinuria which occurs in methionine catabolic pathway.

24-Phenylketonuria can be detected in newborns by screening urine samples with 2,4-Dinitrophenylhydrazone.

25-In diabetic ketosis, liver produces large amounts of ketone bodies, which leads to decrease in pH then impairs tissue function such as central nervous system.

26-In Coris disease, lysosomes become engorged with glycogen because they lack alpha-1,4-glucosidase.

27-In spontaneous depurination, both purines and pyrimidines are stable under normal cellular condition.

28- In methylmalonic acidemia, some patients might respond to the therapy of vitamin B6 to activate the deficient enzyme which called methylmalonyl-CoA mutase.

29- Hyperammonemia is treated by supplementing a protein-restricted diet with large amount of benzoate and phenylacetate.

30- Type 2 DM is a homogeneous syndrome resulting from defects of both insulin secretion and action.

Q2; Choose the ONLY one correct answer for each question.

1- One of the carnitine function in F.A. metabolism is;
   - A- To shuttle long-chain F.A. out of mitochondria
   - B- To shuttle short-chain F.A. out of mitochondria
   - C- To shuttle long-chain F.A. into mitochondria
   - D- To shuttle short-chain F.A. into mitochondria

2- The muscle phosphorylase activity is absent in;
   - A- McArdle glycogen-storage disease (type V)
   - B- Von Gierke glycogen-storage disease (type I)
   - C- Cori glycogen-storage disease (type III)
   - D- Andersen glycogen-storage disease (type IV)

3- The non-sex-linked genes are referred to:
   - A- Dominants
   - B- Autosomal
   - C- Recessive
   - D- Sex-linked

4- Sever hemolytic anemia caused by;
   - A- glycogen-storage disease
   - B- Carnitine deficiency
   - C- G6PD deficiency
   - D- All the above statements are incorrect

5- In germ cell mutation;

A- Causes somatic cell mutation
B- Occurs during mitosis
C- Occurs only in haploid cells.
D- Is known as dominant mutation.

6- There are other metabolic pathways that can generate NADPH in all cells EXCEPT;
A- Hepatocytes
B- Muscle Cells
C- Adipocytes
D- RBC

7- The heterozygous parents have;
A- Each parent has one mutant and one wild-type allele.
B- Mutation in both allele
C- Autosomal recessive trait
D- Abnormal phenotype

8- In Parkinson's disease;
A- Monoamine is precursor of the dopamine
B- Monoamine oxidase is responsible for oxidative deamination of Dopamine
C- Monoamine oxidase is responsible for oxidative deamination and degradation of Dopamine
D- Dopamine oxidase is responsible for oxidative deamination of Dopamine

9- In recessive transmission of enzyme deficiencies;
A- The enzyme is insufficient in heterozygote.
B- The enzyme is insufficient in homozygote mutant.
C- The enzyme is insufficient in both heterozygote and homozygote.
D- The enzyme and its substrate are insufficient in both heterozygote and homozygote.

10- Patients who have Von Gierke disease have an increased dependence on;
A- protein metabolism
B- Fat metabolism
C- Glycogen metabolism
D- All the above are incorrect

11- In "One gene, one enzyme" concept.
A- In mutant gene, the end product will be accumulated.
B- Mutation of a gene leads to deficiency of the substrate.
C- Mutation of a gene leads to deficiency of the enzyme and the substrate.
D- All the above is incorrect

12- One of the primary affected organs in carnitine deficiency is;
A- Liver
B- Adipose tissue
C- Brain

D- Kidney

13- The X-chromosome is important because;
A- It carries genes that are critical for all inherited diseases in human
B- It carries genes affected males than females
C- It carries genes that are critical for human survival
D- Any gene located on this chromosome called X-linked gene

14- GCK enzyme
A- converts glucose to glucose 6- phosphate in pancreatic Beta cells and hepatocytes
B- converts glucose to glucose 6- phosphate in pancreatic Beta cells and adipocytes
C- It's defect causes an increase in glycogen accumulation
D- It's defect related to IDDM

15- The recessive mutation;
A- May leads to gain function
B- May increase the activity of gene product
C- Individual must be a heterozygous
D- Individual must be a homozygous

16- In albinism:
A- The DOPA Quinone is accumulated
B- Both DOPA and DOPA Quinone are deficient
C- Homogentisic acid oxidase will be deficient
E- Phenylalanine will be accumulated

17- The polygenic disorder is
A- Hypolactasia
B- Galactosemia
C- T2DM
D- Carnitine deficiency

18- In dominant and recessive mutation;
A- *A* capital is wild allele in dominant mutation
B- *B* capital is recessive mutant allele
C- In segregation of dominant mutation, *A/a* is mutant
D- In segregation of recessive mutation, *b/B* is mutant