

1. Variant type with most danger sequence:

- a. Benign
- b. Likely benign
- c. Likely pathogenic
- d. Pathogenic
- e. Variant of unknown significance

2. Human mutation characterized by all except:

- a. Mostly neutral
- b. Harmful mutation
- c. Defective protein function mostly
- d. Protect against genetic disorder
- e. Associated with many type of cancer

3. One 15th chromosome larger than usual due to connected to 21 pair, what the type of mutation:

- a. Translocation
- b. Duplication
- c. Insufficiency

4. Mutation has no effect on protein the gene code for, what factor related to that:

- a. Rare mutation
- b. Amino acid have more than 1 codon
- c. Correcting mechanism
- d. A+B
- e. A+B+C

5. Next generation sequence setup require:

- a. Extensive Bioinformatic analysis
- b. Library prepared

- c. A+b true
- d. B+a false

6. Major conclusion of human genome project:

- a. Fewer gene than estimated
- b. Gene encode complex protein
- c. 99% of human genome identical
- d. Contain a lot of repeated
- e. All of above

7. There is 3 alleles locus on c chromosome that control green light sensitivity , if G is normal dominant allele ,g1: a green shift mutation-weakened green sensitivity where only 2 shades are distinguished,g1 is dominant to g2:blindness where red,green,yellow can't be seen !!what is true, they specified it to males only?

- a. half will have green shift mutation, half blind
- b. all blind if it was specified for males
- c. all normal

8. term for condition that affect multi organs

- a. pleiotropy
- b. polygenic inheritance
- c. co dominant
- d. linkage disease
- e. genetic imprinting.

9. Nonsense mutation involve:

- a. Creation of different amino acid
- b. Regulatory sequence
- c. Stop coding

10. Nucleotide repeated sequence 2,3 or 4, 6p:

- a. SNP
- b. Micro stellate

11. Triple repeated, character:

- a. Severity worsen in subsequent generation
- b. Disorder appear to have skipped generation
- c. More likely with paternal aging
- d. Reduced male

12. Feature with marfan syndrome:

- a. PNS tumor & benign
- b. Abnormal heart structure & elongated vessels

13. Cristea site for:

- a. Macromolecule breakdown
- b. Protein synthesis
- c. Oxidation-reduction reaction site
- d. Flevo protein phosphorylation

14. Bill has BOR syndrome, he has bilateral hypoplasia & deaf. His mother mild hearing loss & missing one kidney. Father history of neck cyst resected when was boy? Which genetic concept this case represent?

- a. Pleiotropy
- b. Allele heterogeneity
- c. Heteroplasmy
- d. Variable expressivity

15. to describe disease influenced by genetic and environmental causes , this states:

- a. Multifactorial
- b. polygenic Inheritance

- c. pleiotropy
- d. Co dominant
- e. epigenetic

16. Stepwise of genomic:

- a. Genome sequence, disease causation & treatment
- b. Genome sequence, annotation, variation, genotype, phenotype correlation
- c. None of above

17. Mitochondria is 16659 base pair long, has these gene except:

- a. 22Trna
- b. 2rRNA
- c. 13 protein coding gene
- d. ATPase

18. LHON caused by defect in all of these mitochondrial gene except:

- a. ATP6
- b. ND1
- c. ND4
- d. ND6

19. CORONARY HEART diseases has several genes causing it , this concept is :

- a. polygenic inheritance
- b. epigenetic
- c. Co dominant
- d. multiple alleles

20. an autosomal dominant disease:

- a. Huntington's disease
- b. Duchene
- c. Cystic fibrosis
- d. Tay Sachs

e. hemophilia

21. one of the following Nucleotide transversions is miss matched "not sure of the full question"

- a. C-T
- b. A-T
- c. C-G

22. 2 different mutation that happen on the same locus, the first mutation lead to sickle cell anemia, and the other lead to thalassemia, example of:

- a. Locus mutation
- b. Allele heterogeneity

23. True about hemophilia A:

- a. M more than F
- b. F more than M
- c. Females are the only carrier

Both answers a+b are correct

24. Mitochondrial DNA genome:

Simple ds circular DNA

25. Feature associated with albinism:

Inability to produce sufficient melanin

26. Phenomenon occur when expression of one gene mask or alter expression of another gene:

Epistasis

27. Most commonly accruing variant in human genome:

Single-nucleotide polymorphism

28. Most frequent type of genetic variant:

SNPs

29. Autosomal recessive disease:
Cystic fibrosis

30. Consanguinity associated with:
Autosomal recessive disorder

31. Feature of Duchenne muscular dystrophy:
Muscle weakness & loos coordination

32. 2 different mutation one on FGFR2 and other on FGFR3, both leading to
develop the same disease, example of:
Locus heterogeneity

33. Example of epistasis:
ABO blood group mask expression of MN gene

34. Purpose of x-inactivation in female:
Prevent overexpression of x in female

35. Number of mitochondria in cell depend on
Function state

36. Tay-Sachs is caused by which gene:
HEX A

37. De novo disease
Achondroplasia

38. Regarding autosomal dominant which is wrong:
Only homozygous patients are affected

39. true about Co dominant alleles they were 2 alleles with colors "not sure":
They both contribute in the phenotype was the best choice

40. question about d-loop (was T OR F)

- our exam was 50 question
- ANOTHER left question all were about pedigree and they were easy to answer & solve it

1	D	11	A	21	A
2	D	12	B	22	B
3	A	13	C		
4	B	14	D		
5	C	15	A		
6	E	16	B		
7	B	17	D		
8	A	18	A		
9	C	19	A		
10	B	20	A		