final exam

GENETICS 021

1) An affected man with an X-linked recessive disease could have inherited it from/to:
   a) His son
   b) His brother
   c) His paternal uncle
   d) His maternal uncle

Answer: D

2) The genetic cause of Angelman Syndrome is:
   a) Deletion of 15q11-q13 on maternal chromosome.
   b) Mutation on maternal chromosome.

Answer: B

3) Prader-Willi Syndrome features:
   a) Speech problems
   b) Obesity and hyperphagia

Answer: B

4) A patient has come to the clinic and in diagnosis it appeared that he has lisch nodules and cannot see clearly, which is the diagnosis?

Answer: Neurofibromatosis type 1

5) The tautomeric of cytosine binds to:
   a) Thiamine
   b) Adenine
   c) Guanine
   d) Uracil

Answer: B

6) The type of inheritance when an individual inherits two copies of a chromosome or part of a chromosome from one parent and none from the other parent is called:

a) Uniparental monomy
b) Uniparental disomy

Answer: B

7) Phenylalanine hydroxylase converts:
   a) Phenylalanine to proline.
   b) Phenylalanine to tyrosine.

Answer: B

8) An affected man with cystic fibrosis and his wife is a carrier, what is the chance of them to have a homozygous normal boy?
   a) ¼
   b) 1/16
   c) Zero

Answer: C

9) What is the function of NMD (nonsense mediated decay):
   a) Degradation of truncated protein.
   b) Degradation of premature mRNA.

Answer: B

10) Incomplete dominance is:
   a) Phenotype is intermediate between the phenotype of the two homozygous.
   b) When the phenotype of the heterozygote produce an intermediate product.

Answer: B

11)Tay Sachs characterised by:
Answer: Accumulation of lipid in the brain.

12) What is the difference between germ & somatic cell mutations:
Answer: Germ cell mutations occur in reproductive cells and are inherited, while somatic mutations are not.

13) All true about mutations in regulatory regions except:
Answer: Mutations in noncoding regions does not affect gene expression.

14) Which of the following is Autosomal dominant:
Answer: Huntington disease.

15) What is the difference between mutations and polymorphism:
   a) Mutations are rare and associated with phenotype but polymorphism is common.
   b) Both terms are exchangeable.

c) A mutation is always associated with aggressive diseases and polymorphism is normal.

Answer: A

16) Which class of cystic occur due to splicing defect?

a) Class 1
b) Class 2
c) Class 3
d) Class 4
e) Class 5

Answer: E

17)Tay Sach disease is caused by a mutation in:

Answer: HEXA gene

18) Fragile X syndrome occur in gene:

a) FMR1
b) UBE3A
c) DMBX

Answer: A

19) True about Hemophilia A:

a) More in men compared to women.
b) Female more than male.

Answer: A

20) c.3212_214delinsATCGITCCGGTT means:

a) Deletion of 3 nucleotides and insertion of 12.
b) Deletion of 12 nucleotides and insertion of 12.

Answer: A

21) When there are Many forms of the allele in population but an individual has only , this is called:

Answer: Multiple alleles

22) Not a transversion mutation:

a) G to C
b) T to C
c) A to T

Answer: B

23) Indel means:
a) insertion
b) deletion
c) A + B

Answer: C

24) When Heterozygous are better that homozygous allele, called:
Answer: Over dominance

25) A disease that affects more that one system:
Answer: Pleiropy

26) Nonsense mutations result in:
Answer: Stop codon

27) Which of the following is feature of Neurofibromatosis:
Answer: Fleshy tumors

28) An affected man with X-linked disease and his wife is a carrier, what is the percentage of having a normal daughter and an affected daughter?
a) 50 : 50
b) 0 : 100
c) 100 : 0
d) 2/3 : 1/3

Answer: A

29) AB blood type inheritance:
A) codominance.
B) multiple alleles.
C) complete dominance.

Answer: A

30) The type of dominance in Tay Sachs disease at the molecular level:
Answer: Codominance

31) PKU is characterized by:
Answer: Accumulation of phenylalanine in toxic amounts.

32) Maternally-expressed Imprinted genes constrain growth/proliferation:
A) CDKN1C
B) IGF2

Answer: A

32) Genomic imprinting depends on:
Answer: Methylation

33) Allelic heterogeneity:
A) Different mutations within the same gene lead to a similar disease.
B) Different genes give same clinical features

Answer: A

34) Mutations of different genes produce same disease called:
a) Locus heterogeneity
b) Allelic heterogeneity
C) Pleiotropy
D) Polygenic

Answer: A

35) When a disease is caused by multiple genes then it is:
Answer: polygenic

36) Where does a de novo mutation occur:
A) DNA replication
B) DNA translation
C) DNA repair
D) DNA transcription

Answer: A

37) When a person has a mix of normal and mutated mitochondria, it is called:
A) homoplasmic
B) heteroplasmic
C) homozygous
D) heterozygous

Answer: B

38) When an affected mother give the disease for all of her children, this type of inheritance is called:
A) Mitochondrial
B) Autosomal dominant
C) Autosomal recessive

Answer: A

39) When there is an increase in protein activity, we call it:
A) Hypermorphic
B) Gain of function
C) a+b

Answer: C

40) Which of the following is X-linked recessive:
A) Duchenne syndrome
B) Cystic fibrosis

Answer: A

41) Duchenne muscular dystrophy is caused by:
Answer: Weakness of muscles due to decreased dystrophin.

42) Which of the following nomenclature represents nonsense mutation:
A) p.Trp26Cys
B) p.Trp26*

Answer: B

43) A mutation in DNA coding region .....
A) p.Gly4_Gln6dup
B) c.4375_4385dup

Answer: B

44) BRAF c.1799T>A (p.V600E), Which segment represents the DNA sequence:
A) BRAF
B) c.1799T>A

Answer: B

45) Which mutation of the following cause frameshift:
A) c.212_213insTA
B) pro>Ala

Answer: A

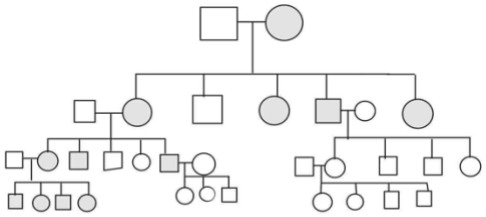
46) Achondroplasia is:
a) AD
b) AR
c) X- linked

Answer: A

47) hypermorphic results in:

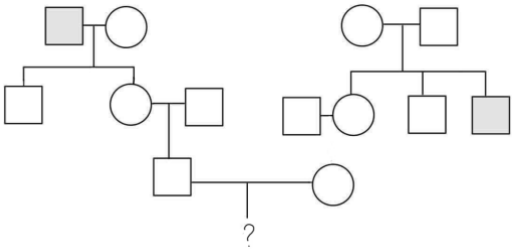
Answer: excess production of protein

48) What is the type of inheritance in this photo:
a) mitochondrial
b) AR
c) X-linked



Answer: B

49) This pedigree shows the inheritance of bloom syndrome, if the last person in the pedigree was a male, what is the percentage of him to have the disease, given that his mother isn't related to his father :
a) 25%
b) 75%
c) 50%
d) Not affected



Answer: D, غالبا انحذف

﴿قُل لا يَستَوِي الخَبيثُ وَالطَّيِّبُ وَلَو أَعجَبَكَ كَثرَةُ الخَبيثِ فَاتَّقُوا اللَّهَ يا أُولِي الأَلبابِ لَعَلَّكُم تُفلِحونَ﴾ [المائدة: ١٠٠]