Please note that the answers provided alongside the questions are examples and may not necessarily be correct. Let me know if you would like any further assistance!

Human genetic variation / clinical variant interpretation

1) Which of the following is true about somatic cells?
    a. They are haploid
    b. They contain 46 chromosomes
    c. They are only found in reproductive organs
    d. They have one pair of sex chromosomes

Answer: B

2) What is the role of genes in inheritance?
    a. They determine physical traits
    b. They code for proteins
    c. They control gene expression
    d. They regulate cellular processes
    e. All the above

Answer: E

3) What is the difference between genetics and genomics?
    a. Genetics studies individual genes, while genomics studies the entire genome
    b. Genetics focuses on inherited traits, while genomics focuses on acquired traits
    c. Genetics deals with somatic cells, while genomics deals with germline cells
    d. Genetics studies animals, while genomics studies plants

Answer: A

4) How many genes are estimated to be present in the human genome?
    a. 10,000
    b. 20,000
    c. 30,000
    d. 40,000

Answer: B

5) Which type of genetic disorder is caused by mutations in a single gene?
    a. Multifactorial inheritance
    b. Monogenic disorder
    c. Chromosomal aberration
    d. Germline mutation

Answer: B

6) In autosomal dominant disorders, who is usually affected?
    a. Homozygotes only
    b. Heterozygotes only
    c. Males only
    d. Both homozygotes and heterozygotes

Answer: D

7) What is the most common type of genetic variation?
    a. Insertions-deletions (indels)
    b. Tandem repeat sequences
    c. Copy number variants (CNVs)
    d. Single nucleotide polymorphisms (SNPs)

Answer: D

8) What is the significance of mutations in genetic disorders?
    a. Most mutations are neutral and have no effect on gene expression
    b. Mutations always lead to genetic disorders
    c. Mutations can cause defective proteins and disrupt normal functions
    d. Mutations only occur during meiosis and have no impact on gene expression

Answer: A

9) How are proteins synthesized from genes?
    a. Through transcription and translation
    b. Through replication and recombination
    c. Through DNA methylation and histone modification
    d. Through gene regulation and epigenetic modifications

Answer: A

10) Which of the following is a type of chromosomal mutation?
    a. Substitution
    b. Insertion
    c. Translocation
    d. Point mutation

Answer: C

11) What is the study of the interactions between genes and the environment called?
    a. Genomics
    b. Genetics
    c. Epigenetics
    d. Molecular biology

Answer: C

12) Which of the following is an example of a recessive genetic disorder?
    a. Huntington's disease
    b. Hemophilia
    c. Down syndrome
    d. Cystic fibrosis

Answer: D

13) What is the name given to the specific location of a gene on a chromosome?
    a. Locus
    b. Allele

c. Exon
d. Intronic region

Answer: A

14) Which of the following is an example of a sex-linked genetic disorder?
a. Sickle cell anemia
b. Duchenne muscular dystrophy
c. Celiac disease
d. Alzheimer's disease

Answer: B

15) What is the term for the occurrence of multiple sets of chromosomes in an organism?
a. Aneuploidy
b. Polyploidy
c. Monosomy
d. Trisomy

Answer: B

16) What is the total number of base pairs in the human mitochondrial genome?
a. 23 pairs
b. 3 billion
c. 16,569
d. Varies between 3-10

Answer: C

17) Which chromosomes are referred to as autosomes?
a. X and Y
b. Chromosomes 1 through 22
c. Circular chromosomes
d. Mitochondrial chromosomes

Answer: B

18) How many genes are there in the human genome approximately?
a. 1,000
b. 10,000
c. 22,000
d. 100,000

Answer: C

19) What is the primary purpose of genetic testing?
a. To determine an individual's ancestry
b. To identify chromosomal abnormalities
c. To rule in/out suspected genetic conditions
d. To detect metabolite levels in the body

Answer: C

20) What is the significance of variant analysis in genetic testing?

a. It helps determine the patient's age
b. It provides information about drug prescriptions
c. It assists in predicting the severity of a disease
d. It identifies the number of mitochondria in cells

Answer: C

21) Which type of genetic testing is performed to detect alterations in a fetus?
a. Carrier testing
b. Newborn screening
c. Prenatal testing
d. Diagnostic testing

Answer: C

22) How are genetic variations classified based on their pathogenicity?
a. Common and rare variants
b. Pathogenic and benign variants
c. Predictive and pre-symptomatic variants
d. Exons and introns

Answer: B

23) Which databases are commonly used for variant annotation and analysis?
a. Genomic Variant Impact and Clinical Variant Interpretation
b. Clinical databases and ACMG guidelines
c. gnomAD browser and Exome Aggregation Consortium
d. Mitochondrial Genome and Human Genome

Answer: C

24) Which type of genetic testing is performed to identify carriers of a recessive genetic condition?
a. Diagnostic testing
b. Pre-symptomatic testing
c. Carrier testing
d. Predictive testing

Answer: C

25) What is the purpose of newborn screening genetic testing?
a. To identify alterations in a fetus
b. To detect alterations in embryos created through in vitro fertilization
c. To identify genetic disorders that can be managed and treated early in life
d. To determine the risk of developing a genetic disorder

Answer: C

26) Pharmacogenetic testing is primarily used for:
a. Identifying genetic disorders in newborns
b. Predicting the severity of a disease
c. Identifying mutations that increase the risk of developing a disorder
d. Identifying the probable individual response to drugs

Answer: D

27) Which type of genetic testing is performed to determine the risk of developing a disorder in a given individual?
    a. Diagnostic testing
    b. Predictive testing
    c. Carrier testing
    d. Forensic testing

Answer: B

28) Preimplantation testing is used to detect genetic changes in embryos created through:
    a. In vitro fertilization
    b. Prenatal testing
    c. Carrier testing
    d. Diagnostic testing

Answer: A

29) Which type of genetic testing is performed to identify an individual for legal or criminal purposes?
    a. Newborn screening
    b. Pre-symptomatic testing
    c. Forensic testing
    d. Prenatal testing

Answer: C

30) What is the purpose of diagnostic genetic testing?
    a. To identify carriers of a recessive condition
    b. To detect alterations in a fetus
    c. To identify or rule out a specific condition
    d. To identify mutations that increase the risk of developing a disorder

Answer: C

31) Which type of genetic testing is performed just after birth to identify genetic disorders?
    a. Carrier testing
    b. Predictive testing
    c. Newborn screening
    d. Pharmacogenetic testing

Answer: C

32) What is the primary purpose of variant calling in genetic testing?
    a. To generate a list of genetic variants associated with a disease
    b. To identify changes in levels of DNA, RNA, chromosomes, and proteins
    c. To determine the risk of developing a genetic disorder
    d. To classify variants into different categories based on their pathogenicity

Answer: A

33) What is the role of bioinformatics in clinical whole genome sequencing?
   a. Preparing patient DNA samples for sequencing
   b. Analyzing genetic variants and their functional impact
   c. Collecting and managing large-scale genetic data
   d. Interpreting clinical reports based on genetic testing results

Answer: B

34) What is the purpose of clinical variant classification?
   a. To identify variants that increase the risk of developing a disorder
   b. To determine the functional impact of genetic variants
   c. To group genetic variants based on their clinical significance
   d. To analyze sequencing data and identify genetic abnormalities

Answer: C

35) How many base pairs are present in mitochondrial DNA (mtDNA)?
   a. 13
   b. 22
   c. 16,569
   d. 37

Mitochondrial diseases

36) How many copies of mtDNA are typically found per mitochondria?
   a. 2-10
   b. 16,569
   c. 37
   d. 1500

B

37) How is mitochondrial DNA inherited?
   a. Paternally inherited
   b. Maternally inherited
   c. Inherited from both parents
   d. Randomly inherited

B

38) How many genes are encoded by mitochondrial DNA?
   a. 13
   b. 22
   c. 37
   d. 1500

c

39) Which of the following is NOT encoded by mitochondrial DNA?
   a. Proteins
   b. Ribosomal RNAs
   c. Transfer RNAs

d. Structural proteins

d

40) True or False: Mitochondrial DNA mutations can occur, and their effects vary based on the location and affected gene.
    a. True
    b. False

a

41) Mitochondrial disorders do not follow the typical Mendelian pattern because:
    a. They are inherited from both parents
    b. They are maternally inherited
    c. They are paternally inherited
    d. They exhibit heteroplasmy

b

42) The organization of the mitochondrial genome is:
    a. Linear and single-stranded
    b. Circular and single-stranded
    c. Linear and double-stranded
    d. Circular and double-stranded

d

43) The non-coding region in the mitochondrial genome that is highly variable and used for population heritage classification is called:
    a. Origin of replication site (OH)
    b. Heavy-strand promoter (HSP)
    c. Light-strand promoter (LSP)
    d. D-loop (displacement loop)

d

44) The genetic code in mitochondria differs from the genetic code in the nucleus in terms of:
    a. Start codons
    b. Stop codons
    c. Introns
    d. Polycistronic mRNAs

b

45) In the mitochondrial genetic code, UGA codes for:
    a. Tryptophan
    b. STOP

c. Arginine
d. Methionine

a

46) Which of the following is true regarding the organization of protein-coding genes in the mitochondrial genome?
a. All protein-coding genes have the same length
b. All protein-coding genes have different sizes and lengths
c. Protein-coding genes are located within the D-loop region
d. Protein-coding genes are intronic

b

47) During oogenesis, the distribution of mitochondrial contents (mutants and normal) in daughter cells is:
a. Selective and controlled
b. Random
c. Predetermined by nuclear DNA
d. Influenced by environmental factors

b

48) Heteroplasmy refers to:
a. The presence of both normal and mutated mtDNA in an individual cell
b. The complete absence of mtDNA in a cell
c. The selective replication of mtDNA in specific tissues
d. The transmission of mtDNA exclusively through the paternal lineage

a

49) The severity of mitochondrial diseases is generally correlated with:
a. The number of mitochondria per cell
b. The percentage of mutant mtDNA molecules in a cell
c. The presence of introns in mtDNA
d. The size of the displacement loop (D-loop)

b

50) The threshold effect in mitochondrial diseases refers to:
a. The dependence of disease severity on nuclear genetics
b. The influence of environmental factors on disease manifestation
c. The different energy needs of tissues and their tolerance for mtDNA mutations
d. The progressive accumulation of mtDNA mutations over time

b

51) . Leber hereditary optic neuropathy (LHON) is caused by mutations in which genes?
a) ND4, ND1, and ND6
b) ND4, ND1, and ND3
c) ND2, ND5, and ND6
d) ND2, ND3, and ND5

a

52) 2. Which complex of oxidative phosphorylation is commonly affected in Leigh syndrome?
a) Complex I
b) Complex II
c) Complex III
d) Complex IV

a

53) 3. Pearson syndrome is associated with a single large-scale deletion in which of the following?

mtDNA

54) 4. MELAS syndrome is caused by mutations in which gene(s)?

a) mtDNA tRNALeu gene

b) mtDNA tRNALys gene

c) mtDNA tRNAMet gene

d) tRNA protein

d

55) 5. Kearns-Sayre Syndrome (KSS) is typically associated with:
a) Inherited mitochondrial DNA mutations

b) Large deletions or duplications of mtDNA

c) Point mutations in mtDNA genes

d) Nuclear DNA mutations

b

56) Which of the following mitochondrial disorders is characterized by the loss of central vision?

a) Leigh syndrome

b) Pearson syndrome

c) MELAS syndrome

d) Leber hereditary optic neuropathy (LHON)

a

57) Kearns-Sayre Syndrome (KSS) is associated with which of the following symptoms?

a) Cardiac abnormalities

b) Progressive hearing loss

c) Digestive problems

d) Short stature

a

58) What is the genetic inheritance pattern of Leber hereditary optic neuropathy (LHON)?

a) Autosomal dominant

b) Autosomal recessive

c) X-linked recessive

d) Maternally inherited

d

5. Pearson syndrome is characterized by which of the following symptoms?

a) Strokes and stroke-like episodes

b) Anemia and lactic acidosis

c) Progressive dementia and hearing loss

d) Ophthalmoplegia and cardiomyopathy

b

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Pharmacogenomics

Genomics is the study of:

A) Individual genes

B) Protein interactions

C) Cellular functions

D) The entire set of genes, their interactions, and functions

Answer: D) The entire set of genes, their interactions, and functions

Pharmacogenomics is a branch of pharmacology that:

A) Studies drug interactions with proteins

B) Focuses on DNA sequencing methods

C) Utilizes DNA to understand drug response and guide drug development

D) Investigates the role of transporters in drug metabolism

Answer: C) Utilizes DNA to understand drug response and guide drug development

Which pathway involves the enzymatic conjugation of drugs or metabolites with hydrophilic compounds?

A) Phase I pathway

B) Phase II pathway

C) Phase III pathway

D) Phase IV pathway

Answer: B) Phase II pathway

CYP450 enzymes are responsible for:

A) Drug excretion by the liver

B) Drug transformation in the kidneys

C) Drug metabolism in the liver

D) Drug transport across cell membranes

Answer: C) Drug metabolism in the liver

Which CYP enzyme is primarily involved in the metabolism of antidepressants and antihypertensive drugs?

A) CYP1

B) CYP2

C) CYP3

D) CYP4

Answer: B) CYP2

Which allele of CYP2D6 is considered the wild type?

A) CYP2D61

B) CYP2D62

C) CYP2D63

D) CYP2D64

Answer: A) CYP2D6*1

TPMT deficiency can lead to severe treatment toxicity when patients are given:

A) Purine analogs

B) Nucleoside analogs

C) Protease inhibitors

D) Antibiotics

Answer: A) Purine analogs

The FDA-approved companion diagnostic for vemurafenib (Zelboraf) is the Cobas 4800 BRAF V600E mutation test, used in treating:

A) Breast cancer

B) Leukemia

C) Melanoma

D) Autoimmune diseases

Answer: C) Melanoma

Which genetic variation is associated with increased risk of hypersensitivity to the drug Abacavir?

A) HLA-B5701 allele

B) HER2 receptor overproduction

C) TPMT deficiency

D) CYP2C92 or CYP2C9*3 alleles

E) Cytochrome P450 CYP2D6 gene deletion

A

Abacavir is a drug used to treat AIDS patients. Which of the following is true about Abacavir?

a. It is a protease inhibitor.

b. It is a fusion inhibitor.

c. It is a nucleoside analog reverse transcriptase inhibitor.

d. It is an integrase inhibitor.

C

What genetic factor is associated with hypersensitivity to Abacavir?

a. HLA-B*5701 allele

b. TPMT deficiency

c. CYP2C9 variant 2 or 3

d. CYP2D6 gene duplication

A

Herceptin (Trastuzumab) is indicated for the treatment of breast cancer patients with:

a. Overproduction of HER2 protein.

b. Underproduction of HER2 protein.

c. Overproduction of TPMT enzyme.

d. Underproduction of CYP2D6 enzyme.

A

What is the main metabolizer of purine analog chemotherapeutic agents?

a. HER2 receptor

b. TPMT enzyme

c. CYP2C9 enzyme

d. CYP2D

B