Introduction of medical genetics

Q1. A researcher finds that a mutation leads to a protein that is misfolded and non-functional. The mutation most likely occurred in which of the following?
A. Promoter region
B. Intron
C. Exon
D. Poly-A tail

Q2. Which of the following codons is a start codon?
A. UGA
B. AUG
C. UAA
D. UAG

Q3. Which enzyme is responsible for synthesizing mRNA from DNA?
A. DNA polymerase
B. RNA polymerase II
C. Ligase
D. Helicase

Q4. A DNA strand has the following sequence: 3'-TACGGTATC-5'. What is the corresponding mRNA sequence?
A. 5'-AUGCCAUAG-3'
B. 5'-ATGCCATAG-3'
C. 3'-AUGCCAUAG-5'
D. 5'-UACGGUAUC-3'

Q5. Which of the following best describes the structure of DNA?
A. Single-stranded molecule with ribose sugar
B. Double helix with base pairing between adenine and guanine
C. Double helix with sugar-phosphate backbone and nitrogenous base rungs
D. Linear molecule with peptide bonds between nitrogenous bases

Q6. A patient is found to have a disorder due to a mutation in mitochondrial DNA. Which of the following inheritance patterns would you expect?
A. Autosomal dominant
B. Autosomal recessive
C. X-linked recessive
D. Maternal inheritance

Q7. Which of the following is **NOT** involved in post-transcriptional modification of mRNA?
A. 5′ cap addition
B. Poly-A tail addition
C. Intron removal
D. Peptide bond formation

Q8. Which type of DNA is used as the template during transcription?

A. Coding (sense) strand
B. Template (antisense) strand
C. mRNA strand
D. tRNA anticodon strand

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Q9.
During which phase of the cell cycle does DNA replication occur?
A. G1
B. G2
C. S phase
D. M phase

Q10.
Which of the following cell types is most likely to remain in G0 phase permanently?
A. Hepatocytes
B. Neurons
C. Skin epithelial cells
D. Intestinal crypt cells

Q11.
A cell completes DNA replication but fails to detect damage before mitosis. This checkpoint failure occurred in which phase?
A. G1
B. S
C. G2
D. M

Q12.
Which phase of mitosis is characterized by alignment of chromosomes at the cell's equator?
A. Prophase
B. Metaphase
C. Anaphase
D. Telophase

Q13.
Which protein complex is responsible for cell cycle progression through checkpoints?
A. DNA polymerase
B. Ribosomes
C. Cyclin-CDK complex
D. tRNA synthetase

Q14.
A drug inhibits mitotic spindle formation. Which phase of mitosis is directly affected?
A. Anaphase
B. Prophase
C. Metaphase
D. Cytokinesis

Q15.
A mutation in the promoter region of a gene is most likely to affect which of the following processes?
A. Protein folding
B. Translation termination
C. Binding of RNA polymerase
D. Ribosome assembly

Q16
All of the following statements about the human genome are true **EXCEPT**:

a. The genome includes all the DNA in an organism
b. The Human Genome Project began in 1990
c. The complete DNA sequence was finished in 2003
d. The genome contains only genes that make proteins

Q17.
What is the basic unit of inheritance that is a segment within a long strand of DNA with specific instructions for the production of one specific protein?
a. Chromosome
b. Genome
c. Allele
d. Gene

Q18
A 14-year-old girl presents for evaluation due to delayed puberty. Physical examination shows no breast development and no pubic or axillary hair. Laboratory studies reveal

elevated levels of follicle-stimulating hormone (FSH) and luteinizing hormone (LH), and a karyotype analysis reveals a total of 45 chromosomes.

Which of the following best explains the chromosomal basis of this patient's condition?

a. Failure of homologous chromosomes to separate during meiosis I
b. Mitotic nondisjunction in somatic cells after fertilization
c. Inheritance of two X chromosomes from the mother
d. Loss of a chromosome during anaphase of mitosis
e. Crossing over failure during prophase I of meiosis

Explanation
The patient's features suggest **Turner syndrome (45,X)**, which results from nondisjunction—an error during meiosis, most often in the paternal gamete. Normally, somatic cells are diploid (46 chromosomes), with homologous autosomes and one pair of sex chromosomes. In meiosis I, homologous chromosomes are supposed to separate. Failure to do so can lead to a gamete lacking a sex chromosome, which, when fertilized by a normal gamete, results in monosomy (45,X).

Q19 . What is the **purpose of meiosis** in humans?
→ To reduce diploid cells (2n) into haploid gametes (n) and generate genetic diversity.

Q20. In which stage of meiosis does **crossing over** occur?
→ Prophase I

Q21. What is the name of the structure where crossing over happens?
→ Chiasma (plural: **chiasmata**)

Q22.
A geneticist is analyzing gametes from a meiosis experiment. She notes that homologous chromosomes exchanged segments of DNA early in division. Which of the following stages did this occur in?
A. Metaphase I
B. Anaphase I
C. Prophase I
D. Telophase II

Explanation: Crossing over occurs during Prophase I, at chiasmata between homologous chromosomes.

Q23.
Which of the following best explains why sister chromatids are not genetically identical after meiosis?
A. Errors in DNA polymerase
B. Lack of telomerase
C. Crossing over during Prophase I
D. Unequal cytoplasmic division

Explanation: Because of chiasma formation, exchanged segments result in unique combinations of alleles.

Q24.

In which of the following phases are homologous chromosomes separated, while sister chromatids remain intact?

A. Anaphase I
B. Anaphase II
C. Metaphase I
D. Telophase II

High-Yield Clues for Exams

Prophase I = Crossing over → chiasmata

Anaphase I = Homolog separation (not sister chromatids)

Anaphase II = Sister chromatids separate

Genetic variation is due to:

Crossing over (Prophase I)

Random alignment (Metaphase I)

(**Straight Facts**):

Q1. How many functional sperm cells are formed from one spermatogonium?
→ 4 haploid sperm cells

Q2. At what stage are female primary oocytes arrested at birth?
→ Prophase I

Q3. What is the trigger for completion of Meiosis II in the secondary oocyte?
→ Fertilization by a sperm cell

Q4. In which gametogenesis process is cytoplasm divided unequally?
→ Oogenesis (ovum gets most of it; polar bodies get very little)

Q5. Which type of division occurs during Meiosis II?
→ Equational division (sister chromatids separate)

Q25.

A diploid spermatogonium undergoes complete spermatogenesis. How many single-chromatid cells will result?

A. 1
B. 2

C. 3
D. 4
Explanation: One spermatogonium produces 4 spermatids (haploid, single-chromatid) after Meiosis II.

Q26.
In females, when does the primary oocyte resume meiosis after being arrested in Prophase I?
A. At birth
B. At fertilization
C. At ovulation
D. At puberty
Explanation: Meiosis I completes at ovulation, producing a secondary oocyte and a polar body.

Q27.
A 13-year-old girl ovulates for the first time. Which cells result from the first meiotic division of her primary oocyte?
A. Two equal-sized secondary oocytes
B. One secondary oocyte and one polar body
C. Two polar bodies
D. One ovum only
Explanation: Meiosis I in females is asymmetric, producing one large cell and one small polar body.

Q28.
A fertilized secondary oocyte completes Meiosis II. What is the genetic content of the resulting ovum?
A. Diploid
B. Haploid, 46 chromatids
C. Haploid, 23 chromatids
D. Diploid, 92 chromatids
Explanation: Meiosis II separates sister chromatids → haploid ovum with 23 single chromatids.

Q29
Which of the following best describes the difference in cytoplasmic division between spermatogenesis and oogenesis?
A. Equal in both
B. Equal in males, unequal in females
C. Equal in females, unequal in males
D. Unequal in both
Explanation: Males produce equal-sized sperm; females produce one ovum with most cytoplasm and 2–3 polar bodies.

Q30
Which of the following best explains why nondisjunction during meiosis I results in more severe chromosomal abnormalities than in meiosis II?

A. Chromosome duplication
B. All resulting gametes are aneuploid
C. Both homologs end up in the same gamete
D. It only occurs in oogenesis

Q31.
A newborn presents with hypotonia, upslanting palpebral fissures, and a single palmar crease. Cytogenetics reveals 47 chromosomes. This condition is most likely caused by:
A. Deletion during mitosis
B. Nondisjunction during meiosis I
C. Translocation during fertilization
D. Somatic mutation post-fertilization

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