Genetics and Genomics in Medicine Chapter 1

Questions & Answers

Multiple Choice Questions

Question 1.1

In a DNA double helix each type of base forms a stable base pair with only one type of base. When bases on an RNA strand are involved in base pairing, which bases can form stable base pairs with more than one type of base?

a) Adenine and uracil
b) Cytosine and uracil
c) Guanine and uracil
d) Uracil

Answer 1.1

c) Guanine and uracil

Explanation 1.1

Guanine and uracil can form G-U base pairs in addition to G-C and U-A base pairs.

Question 1.2

Regarding the structure of bases in nucleic acids, which, if any, of the following statements is incorrect?

a) Pyrimidines have a single heterocylic ring consisting of four carbon atoms plus two nitrogen atoms that are linked to the same carbonyl (-C=O) group
b) Purines have two heterocyclic rings, each with two nitrogen atoms
c) Thymine = 5-methyluracil.
d) Purines carry an amino group, unlike pyrimidines

Answer 1.2

d) Purines carry an amino group, unlike pyrimidines

Explanation 1.2

Cytosine is a pyrimidine in which an amino group is attached to carbon atom 4.

Question 1.3

Regarding the sugar residues in nucleic acids, which, if any, of the following statements is incorrect?

a) The sugar is always based on a heterocyclic ring that consists of one oxygen atom and four carbon atoms, one of which is connected to a fifth carbon atom, the 5' carbon atom.
b) The internal sugar residues in a DNA strand normally have free hydroxyl groups, unlike those in an RNA strand.
c) Bases are covalently linked to the carbon 1' atoms of the sugars.
d) Neighboring sugar residues are linked to each other by a phosphodiester bond that connects the 3' carbon atom on one sugar to the 5' carbon atom of its neighbour.

Answer 1.3

b) The internal sugar residues in a DNA strand normally have free hydroxyl groups, unlike those in an RNA strand.

Question 1.4

Regarding DNA replication, which, if any, of the following statements is incorrect?

a) DNA replication is said to be semi-conservative because for each DNA helix only one strand serves as a template for new DNA synthesis.
b) The two DNA strands of the parental DNA helix are unwound using a DNA helicase.
c) The replication fork marks the point at which DNA is actively being replicated from a single DNA duplex to form new DNA strands.
d) The synthesis of new DNA strands always proceeds in the 5' → 3' direction.

Answer 1.4

a) DNA replication is said to be semi-conservative because for each DNA helix only one strand serves as a template for new DNA synthesis.

Explanation 1.4

During DNA replication, both DNA strands of the parental double helix are simultaneously replicated.

Question 1.5

Regarding DNA replication and DNA synthesis, which, if any, of the following statements is incorrect?

a) The leading strand is replicated continuously.
b) The lagging strand is replicated discontinuously: the new DNA is synthesized as short pieces of DNA that must be ligated together.

c) A DNA polymerase always requires a DNA template to synthesize a complementary DNA.
d) A DNA polymerase requires a short RNA primer when synthesizing new DNA strands using a DNA template.

Answer 1.5

c) A DNA polymerase always requires a DNA template to synthesize a complementary DNA.

Explanation 1.5

Some types of DNA polymerase, known as reverse transcriptases, naturally use RNA as templates for synthesizing DNA.

Question 1.6

Which is the best definition of noncoding RNA?

a) A mutant mRNA that is not capable of making a protein.
b) Any RNA sequence that is not translated to make a protein
c) Any RNA sequence in an mRNA, or in the primary transcript used to make the mRNA, that is not translated to make a protein.
d) A functional RNA that is not translated to make a protein.

Answer 1.6

d) A functional RNA that is not translated to make a protein.

Explanation 1.6

Although b) might appear to be an acceptable definition it would include short-lived sequences such as transcribed introns, and so on, and so noncoding RNA is usually taken to mean a functional RNA that is not translated.

Question 1.7

If we take the number of chromosomes in a haploid sperm cell or egg cell to be *n* and the total content of genomic DNA to be C, what is the number of chromosomes and the total genomic DNA content of a diploid G_1 phase cell and in a diploid G_2 phase cell from the same organism.

a) *n* chromosomes, DNA content of 2*C* in G_1; 2*n* chromosomes, DNA content of 4*C* in G_2.
b) 2*n* chromosomes, DNA content of 2*C* in G_1; 4*n* chromosomes, DNA content of 4*C* in G_2.
c) 2*n* chromosomes, DNA content of 2*C* in G_1; 4*n* chromosomes, DNA content of 2*C* in G_2.
d) 2*n* chromosomes, DNA content of 2*C* in G_1; 2*n* chromosomes, DNA content of 4*C* in G_2.

Answer 1.7

d) 2*n* chromosomes, DNA content of 2*C* in G_1; 2*n* chromosomes, DNA content of 4*C* in G_2.

Question 1.8

Regarding the cell cycle, which, if any, of the following statements is correct?

a) The M phase and the S phase are the critically important phases of the cell cycle; according to circumstance, the G_2 phase and the G_1 phase are dispensable.
b) The M phase is the only phase in the cell cycle when chromosomes stained with chemical dyes are visible under the light microscope.
c) The G_0 phase is a specialized phase that allows terminally differentiated cells to irretrievably exit the cell cycle and become post-mitotic cells.
d) The G_0 phase is a modified version of the G_2 phase.

Answer 1.8

b) The M phase is the only phase in the cell cycle when chromosomes stained with chemical dyes are visible under the light microscope.

Fill in the Blanks Questions

Question 1.9

Fill in the numbered blanks with single words.

When one of our chromosomes undergoes replication, the very long double helix within the chromosome DNA must also _____1_____. To do that the DNA double helix must be unwound using a dedicated enzyme known as a ____2_____. The individual parental DNA strands serve as ____3_____ for new DNA synthesis to generate new ____4____ daughter DNA strands. Because each parental double helix generates two daughter double helixes that contain one parental strand plus one newly synthesized DNA strand, DNA replication is said to be _____5_____.

Answer 1.9

1. replicate. 2. helicase. 3. templates. 4. complementary. 5. semi-conservative.

Question 1.10

Fill in the numbered blanks with single words or numbers.

Different types of DNA polymerase work in our cells, and in each case the new DNA strand is synthesized in the ___1___ prime to ____2___ prime direction. Most of our DNA polymerases use a DNA template from which a complementary new DNA strand is made. To help initiate DNA synthesis, a DNA polymerase requires an _____3____ primer that must first be synthesized by an _____3____ polymerase. A few DNA polymerases , known as ____4____ _____5_____ , use an _____3____ template to synthesize DNA. For example, an enzyme known as _____6_____ uses an _____3____ template to replicate DNA at the very ends of our chromosomes.

Answer 1.10

1. 5. 2. 3. 3. RNA. 4. reverse. 5. transcriptases. 6. telomerase.

Question 1.11

Fill in the numbered blanks with single words.

When a DNA double helix is replicated, the point at which active DNA replication is occurring (where two double helices are being generated from one parental double helix) is known as a ____1____ ____2____. As DNA synthesis occurs, one of the DNA strands that is being

produced, known as the ____3_____ ____4_____ , is synthesized continuously in the same direction as the movement of the ____1____ ____2____. But the other strand, known as the ____5_____ ____4_____ is synthesized in the opposite direction to the movement of the ____1____ ____2____. As a result it must be synthesized in short pieces (known as ____6_____ fragments, each synthesized using an RNA ____7____); each RNA_____7_____ binds to the DNA template a short distance in advance, allowing DNA synthesis in the backward direction. As a result of this asymmetry in DNA synthesis, DNA replication is said to be semi-____8_____.

Answer 1.11
1. replication. 2. fork. 3. leading. 4. strand. 5. lagging. 6. Okazaki. 7. primer. 8. discontinuous.

Question 1.12

Fill in the numbered blanks below using single words or symbols.

A chromosome has one long DNA molecule. When a chromosome replicates, it forms two ____1_____ ____2_____, each containing a copy of the original DNA molecule, that are held together initially across their lengths by multi-subunit protein complexes called ____3_____. At a later stage, most of the ____3_____ are removed but some remain at the centromere to keep the two ____1_____ ____2_____ together until the _____4_____ stage of mitosis. At this point the remaining ____3____ begin to be removed so that the ____1_____ ____2_____ dissociate to become separate chromosomes. The _____5_____ attached to the _____6_____ at the centromeres of the two chromosomes ensure that the two chromosome copies migrate to opposite poles of the dividing cell (the identical chromosome copies normally ____7_____ equally into the daughter cells).

Answer 1.12
1. sister. 2. chromatids. 3.cohesins. 4. anaphase. 5. microtubules. 6. kinetochore. 7. segregate.

Question 1.13

Fill in the numbered blanks below using single words.

____1_____ is the usual form of cell division and occurs after a previous round of chromosome (and DNA) ____2_____. The point of this type of cell division is to allow an increase in cell ___3____. As might be expected, ____1____ is particularly prevalent during periods of growth, but even in a fully grown adult there is a need for cell divisions in order to replace certain cells that have ___4____ lifespans, such as ___5____ cells and some types of epithelial cells, notably those in the _____6_____ tract.

Answer 1.13
1. mitosis. 2. replication. 3. number. 4.short. 5. blood. 6. gastrointestinal.

Question 1.14
Fill in the numbered blanks below using single words.

____1_____ is a specialized type of cell division that is exclusively used to produce ____2_____, and so occurs only in cells required for that purpose in the ____3_____ and ____4____. It really comprises two successive cell divisions following a single round of DNA synthesis. As a result, what was a single diploid germ cell precursor cell gives rise to ____5____ haploid ___6____ cells in men. In women, however, the two cell divisions are each asymmetric, producing one large cell plus a smaller cell (called a _____7_____ _____8_____) that is discarded, and so ultimately generating just a single ____9_____ cell.

Answer 1.14
1. meiosis. 2. gametes. 3. testis. 4. ovary. 5. four. 6. sperm. 7. polar. 8. body. 9. egg.

Question 1.15
Fill in the numbered blanks below using single words

The whole point of meiosis is to increase genetic ___1___ . That occurs in two ways. First, as a result of independent ___2____ of homologous chromosomes, sperm and egg cells contribute only one of the two alleles at each diploid locus in the two parents. Secondly, the process of ____3_____ ensures that the individual chromosomes in a sperm or egg cell are a mixture of alleles present on both homologs of the paternal or maternal grandparents. _____3_____ involves breakage and re-joining of DNA strands on _____4_____ chromatids that can be observed under the microscope. The point of ___5____ (at which the chromatids have broken and re-joined) can be visualized under the microscope, when it is referred to as a _____6____ .

Answer 1.15
1. variation (or diversity). 2. assortment. 3. recombination. 4. non-sister. 5. crossover. 6. chiasma.

Question 1.16
Fill in the numbered blanks below.

The ends of linear chromosomes have specialized nucleoprotein structures known as ____1_____. The DNA component of a ____1_____ consists of short ____2____ repeats that have been highly ____3_____ during evolution. In humans and other vertebrates the DNA repeat

is a hexanucleotide, with the sequence ____4_____. As a result, one of the two DNA strands is rich in guanines (G-rich) and one that is rich in cytosines (C-rich). At the very end of the ___1____ DNA, the G-rich strand has about 30 repeats that are ____5____. This short ____5____ overhang is able to bend backwards to base pair with complementary repeat sequences on the C-strand (after displacing the G-rich strand over a short region). The resulting structure, known as a ___6____ protects the ____1____ DNA from natural cellular _____7_____ that naturally remove some nucleotides from broken DNA strands during DNA repair.

Answer 1.16

1. telomere(s). 2. tandem. 3. conserved. 4. TTAGGG. 5.single-stranded. 6. T-loop. 7. exonucleases.

Essay & Listing Questions

Question 1.17

The chromosomes of our cells contain immensely long DNA molecules. Describe how they are compacted (a) in an interphase cell and (b) in preparation for cell division. By how much is the linear length of the DNA compacted in these two situations?

Answer 1.17

a) In an interphase cell the 2-nm-thick DNA double helix is compacted by a factor of about 50. It is periodically coiled round complexes of eight histone proteins to form a filament studded with 10-nm-thick nucleosomes and then is further coiled to form a 30 nm-thick chromatin fiber (Figure 1.8, page 8).
b) In preparation for cell division, the chromatin fiber is further coiled in various ways including the formation of looped domains. The result is that the linear length of the double helix is compacted by a factor of about 10,000.

Question 1.18

What is meant by ploidy? Give naturally occurring examples of (a) a haploid cell (b) a diploid cell (c) a polyploid cell (d) a nulliploid cell.

Answer 1.18

Ploidy means the number of copies of the *chromosome set* (the *different* chromosomes in a cell). Examples are:

a) haploid: sperm and egg cells;
b) diploid: the zygote, and most somatic cells such as lymphocytes, neurons and so on;
c) polyploid: muscle fiber cells (which have multiple nuclei) and megakaryocytes where the chromosomes have replicated several times without an intervening cell division;
d) nulliploid: erythrocytes (lack a nucleus) and terminally differentiated skin cells (lack organelles including a nucleus).

Explanation 1.18

See Box 2.2 on page 29.