## Genetics and Genomics in Medicine Chapter 1 Questions

## 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

## 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 $(-\mathrm{C}=\mathrm{O})$ group
b) Purines have two heterocyclic rings, each with two nitrogen atoms
c) Thymine $=5$-methyluracil.
d) Purines carry an amino group, unlike pyrimidines

## 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.

## 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 $5^{\prime} \rightarrow 3^{\prime}$ direction.

## 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.

## 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.

## 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 $\mathrm{G}_{1} ; 2 n$ chromosomes, DNA content of $4 C$ in $\mathrm{G}_{2}$.
b) $2 n$ chromosomes, DNA content of $2 C$ in $\mathrm{G}_{1} ; 4 n$ chromosomes, DNA content of $4 C$ in $\mathrm{G}_{2}$.
c) $2 n$ chromosomes, DNA content of $2 C$ in $\mathrm{G}_{1} ; 4 n$ chromosomes, DNA content of $2 C$ in $\mathrm{G}_{2}$.
d) $2 n$ chromosomes, DNA content of $2 C$ in $\mathrm{G}_{1} ; 2 n$ chromosomes, DNA content of $4 C$ in $\mathrm{G}_{2}$.

## 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 $\mathrm{G}_{2}$ phase and the $\mathrm{G}_{1}$ 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 $\mathrm{G}_{0}$ phase is a specialized phase that allows terminally differentiated cells to irretrievably exit the cell cycle and become post-mitotic cells.
d) The $\mathrm{G}_{0}$ phase is a modified version of the $\mathrm{G}_{2}$ phase.

## 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 $\qquad$ 1 $\qquad$ . 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 $\qquad$ for new DNA synthesis to generate new $\qquad$ 4 $\qquad$ 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
$\qquad$ 5 $\qquad$ .

## 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 $\qquad$ 3 $\qquad$ primer that must first be synthesized by an $\qquad$ 3 ___ polymerase. A few DNA polymerases , known as $\qquad$ 4 $\qquad$
$\qquad$ 5 $\qquad$ , use an $\qquad$ 3 template to synthesize DNA. For example, an enzyme known as $\qquad$ uses an $\qquad$ 3 template to replicate DNA at the very ends of our chromosomes.

## 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
$\qquad$ 1 ___ 2 $\qquad$ . As DNA synthesis occurs, one of the DNA strands that is being produced, known as the $\qquad$ 3 $\qquad$ 4 $\qquad$ , is synthesized continuously in the same direction as the movement of the $\qquad$ $1 \quad 2$ 2 . But the other strand, known as the
$\qquad$ 5 $\qquad$ 4 $\qquad$ is synthesized in the opposite direction to the movement of the - $\qquad$ . As a result it must be synthesized in short pieces (known as $\qquad$ 6 $\qquad$ fragments, each synthesized using an RNA $\qquad$ 7 $\qquad$ ); each RNA $\qquad$ 7 $\qquad$ 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-
$\qquad$
8 $\qquad$ .

## 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
$\qquad$ $1 \_2$ $\qquad$ , each containing a copy of the original DNA molecule, that are held together initially across their lengths by multi-subunit protein complexes called $\qquad$ 3 $\qquad$ . At a later stage, most of the $\qquad$ 3 $\qquad$ are removed but some remain at the centromere to keep the two $\qquad$ 1 __ 2 2 __ together until the $\qquad$ stage of mitosis. At this point the remaining $\qquad$ 3 begin to be removed so that the $\qquad$ 1 2 $\qquad$ dissociate to become separate chromosomes. The $\qquad$ 5 $\qquad$ attached to the $\qquad$ 6 $\qquad$ 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 $\qquad$ 7 $\qquad$ equally into the daughter cells).

## Question 1.13

Fill in the numbered blanks below using single words.
$\qquad$
$\qquad$ is the usual form of cell division and occurs after a previous round of chromosome (and DNA) $\qquad$ . The point of this type of cell division is to allow an increase in cell
$\qquad$
$\qquad$ . As might be expected, $\qquad$ 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 ___ $4 \ldots \quad$ lifespans, such as ___ $5 \ldots \quad$ cells and some types of epithelial cells, notably those in the $\qquad$ tract.

## Question 1.14

Fill in the numbered blanks below using single words.
$\qquad$
1 is a specialized type of cell division that is exclusively used to produce
$\qquad$ , and so occurs only in cells required for that purpose in the $\qquad$ 3 $\qquad$ 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 $\qquad$ 5 5 haploid ___ _ cells in men. In women, however, the two cell divisions are each asymmetric,
producing one large cell plus a smaller cell (called a $\qquad$ 7 $\qquad$ 8 $\qquad$ ) that is discarded, and so ultimately generating just a single $\qquad$ 9 cell.

## Question 1.15

Fill in the numbered blanks below using single words

The whole point of meiosis is to increase genetic $\qquad$ 1 $\qquad$ . That occurs in two ways. First, as a result of independent $\qquad$ 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
$\qquad$ 3 $\qquad$ 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. $\qquad$ 3 involves breakage and re-joining of DNA strands on $\qquad$ 4 $\qquad$ chromatids that can be observed under the microscope. The point of $\qquad$ 5 $\qquad$ (at which the chromatids have broken and re-joined) can be visualized under the microscope, when it is referred to as a $\qquad$ 6 $\qquad$ .

## Question 1.16

Fill in the numbered blanks below.

The ends of linear chromosomes have specialized nucleoprotein structures known as
$\qquad$
1 $\qquad$ . The DNA component of a $\qquad$ 1 $\qquad$ consists of short $\qquad$ 2 $\qquad$ repeats that have been highly ___ 3 $\qquad$ during evolution. In humans and other vertebrates the DNA repeat is a hexanucleotide, with the sequence $\qquad$ 4 $\qquad$ . 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
$\qquad$
$\qquad$ DNA, the G-rich strand has about 30 repeats that are $\qquad$ 5 $\qquad$ . This short
5 $\qquad$ 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 ___ that naturally remove some nucleotides from broken DNA strands during DNA repair.

## 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?

## 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.

