Chromosome Mutations: Variation in Number and Arrangement

1) The condition that exists when an organism gains or loses one or more chromosomes but not a complete haploid set is known as _____.

A) polyploidy
B) euploidy
C) aneuploidy
D) triploidy
E) trisomy

2) Trisomy 21, or Down syndrome, occurs when there is a normal diploid chromosomal complement but one (extra) chromosome 21. While there is reduced fertility in both sexes, females have higher fertility than males. Van Dyke et al. (1995; *Down Syndrome Research and Practice* 3(2):65—69) summarize data involving children born of Down syndrome individuals. Given the fact that conceptuses with 48 chromosomes (four #21 chromosomes) are not likely to survive early development, what percentage of surviving offspring would be expected to have Down syndrome if both parents have Down syndrome?

A) One-third of the surviving offspring would be expected to have Down syndrome.
B) All the children would be expected to have Down syndrome.
C) None of the surviving offspring would be expected to have Down syndrome.
D) Two-thirds of the surviving offspring would be expected to have Down syndrome.
E) One-half of the surviving offspring would be expected to have Down syndrome.

3) Although the most frequent forms of Down syndrome are caused by a random error, nondisjunction of chromosome 21, Down syndrome occasionally runs in families. The cause of this form of familial Down syndrome is _____.

A) an inversion involving chromosome 21
B) a chromosomal aberration involving chromosome 1
C) too many X chromosomes
D) a translocation between chromosome 21 and another acrocentric chromosome
E) a maternal age effect

4) The condition known as *cri-du-chat* syndrome in humans has a genetic constitution designated as _____.

A) 45, X
B) heteroplasmy
C) 46, 5p-
D) triploidy
E) trisomy

5) What explanation is generally given for lethality of monosomic individuals?

6) In what way might gene duplication play a role in evolution?

7) Name the polyploid condition that is formed from the addition of an extra set of chromosomes identical to the normal diploid complement of the same species.

8) What is the chromosome number for each of the following conditions:

A) Turner syndrome

B) Klinefelter syndrome

C) Down syndrome

9) Under what circumstance can an individual with Down syndrome have 46 chromosomes?

10) The term *aneuploidy* is synonymous with the term *segmental deletion*. True or false?

11) Nondisjunction is viewed as a major cause of aneuploidy. True or false?

12) Assume that a species has a diploid chromosome number of 24. The term applied to an individual with 25 chromosomes would be *triploid*. True or false?

13) An individual with Patau syndrome would be called a triploid. True or false?

14) Assume that an organism has a diploid chromosome number of 14. There would be 28 chromosomes in a tetraploid. True or false?

15) A paracentric inversion is one whose breakpoints do not flank the centromere. True or false?

16) In general, inversion and translocation heterozygotes are as fertile as organisms whose chromosomes are in the standard arrangement. True or false?

Mutation and DNA Repair

1) Which of the following class of mutations can result in multiple contiguous amino acid changes in proteins?

A) base analog
B) single nucleotide variant
C) inversion
D) indel
E) translocation

2) Mutations that arise in nature, from no particular artificial agent, are called _____.

A) oblique mutations
B) induced mutations
C) spontaneous mutations
D) chromosomal aberrations
E) cosmic mutations

3) Which of the following mutagens are classified as base analogs?

A) acridine orange and proflavine
B) UV-B
C) ultraviolet light and cosmic radiation
D) 5-bromouracil and 2-amino purine
E) hydroxyurea and peroxidase

4) Transposons, or jumping genes, are DNA elements that move within the genome. In which organismic groups are transposons found?

A) bacteria
B) eukaryotes
C) mammals
D) ancient bacteria
E) all of the above

5) Mutations may exert a variety of effects on living systems. List at least three categories of mutations based on their biological effects.

6) Apurinic sites (AP sites) involve a spontaneous loss of a(n) _____ in an intact double-helix DNA molecule.

7) Describe the mutagenic action of the following two mutagens: 5-bromouracil and ultraviolet light.

8) Considering the electromagnetic spectrum, identify likely mutagens from the following list: radio waves, microwaves, infrared, ultraviolet, X-rays, gamma rays, cosmic rays.

9) What is meant by the term *photoreactivation* repair?

10) The process of error correction of mismatched bases carried out by DNA polymerases is called _____.

11) Loss-of-function mutations eliminate the function of a gene product and may be dominant or recessive. True or false?

12) Of the two cell lines that can contain a mutation in an organism, somatic and germ line, the latter is more consequential to subsequent generations. True or false?

13) The shorter the wavelength of a radiation source, the greater its likelihood of causing damage. True or false?

14) A missense mutation causes premature chain (protein) termination. True or false?

Cancer and Regulation of the Cell Cycle

1) Driver mutations provide a growth advantage to a tumor cell. Which type of mutation is known to accumulate in cancer cells but has no direct contribution to the cancer phenotype?

A) alteration mutations
B) passenger mutations
C) carrier mutations
D) indirect mutations
E) insignificant mutations

2) The retinoblastoma protein (pRB), like p53, serves as a(n) _____ in regulating the cell cycle.

A) tumor suppressor
B) tumor enhancer
C) up regulator
D) oncogene
E) pseudo-oncogene

3) Mutant versions of genes that are normally involved in promoting the cell cycle are known as _____.

A) tumor suppressors
B) proto-oncogenes
C) oncogenes
D) malignant genes
E) attenuators

4) In sporadic cases of retinoblastoma, how many gene mutations are thought to be necessary in the same cell for a tumor to develop?

A) one
B) two
C) four
D) six
E) There is insufficient information to answer this question.

5) Chronic myelogenous leukemia appears to be associated with a chromosomal rearrangement. How is a chromosomal rearrangement responsible for this disease?

6) Which three stages or transitions in the cell cycle seem to serve as points of control (checkpoints)?

7) Describe the cellular and molecular function of the *ras* gene family and the consequences of mutations in *ras*.

8) What is the name of a normal gene that serves to promote cellular division?

9) What are two properties shared by all types of cancer?

10) Name three human cancers with a genetic predisposition. What appears to be the genetic cause of each?

11) List at least three environmental agents or factors that are known to cause cancer.

12) Any agent that causes damage to DNA is a potential carcinogen. True or false?

13) There are several checkpoints in the mitotic cell cycle. All occur in the S phase. True or false?

14) The gene p53 is called the "guardian of the genome" because it corrects mutations in the spindle apparatus before nondisjunction can occur. True or false?

15) A *tumor-suppressor gene* normally functions to limit cell replication. True or false?