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
Answer: C

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.
Answer: D

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
Answer: D

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
Answer: C

5) What explanation is generally given for lethality of monosomic individuals?
Answer: Monosomy may unmask recessive lethals that are tolerated in heterozygotes carrying the wild-type allele.

6) In what way might gene duplication play a role in evolution?
Answer: Gene duplication provides a way in which new genes arise. By duplicating a gene, the duplicated copy or the original gene is able to mutate without necessarily having an adverse influence on the phenotype.

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.
Answer: autotetraploidy, assuming the normal chromosome complement is diploid

8) What is the chromosome number for each of the following conditions:
Turner syndrome
Klinefelter syndrome
Triploid
Down syndrome
Answers:

Turner syndrome (female, no Barr bodies)	45
Klinefelter syndrome (male, one Barr body)	47
Triploid	69
Down syndrome (trisomic)	47

9) Under what circumstance can an individual with Down syndrome have 46 chromosomes?
Answer: if he or she carries a Robertsonian translocation, 14/21, for example

TRUE or FALSE

10) The term *aneuploidy* is synonymous with the term *segmental deletion*.
Answer: FALSE

11) Nondisjunction is viewed as a major cause of aneuploidy.
Answer: TRUE

12) Assume that a species has a diploid chromosome number of 24. The term applied to an individual with 25 chromosomes would be *triploid*.
Answer: FALSE

13) An individual with Patau syndrome would be called a triploid.
Answer: FALSE

14) Assume that an organism has a diploid chromosome number of 14. There would be 28 chromosomes in a tetraploid.
Answer: TRUE

15) A paracentric inversion is one whose breakpoints do not flank the centromere.
Answer: TRUE

16) In general, inversion and translocation heterozygotes are as fertile as organisms whose chromosomes are in the standard arrangement.
Answer: 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 change
C) inversion
D) frameshift
E) translocation
Answer: D

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
Answer: C

3) Which of the following mutagens are classified as base analogs?
A) acridine orange and proflavine
B) ethylmethane sulfonate and ethylmethylketone peroxide
C) ultraviolet light and cosmic radiation
D) 5-bromouracil and 2-amino purine
E) hydroxyurea and peroxidase
Answer: D

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
Answer: E

5) Mutations may exert a variety of effects on living systems. List at least three categories of mutations based on their biological effects.
Answer: morphological mutations, nutritional or biochemical mutations, behavioral mutations, regulatory mutations, lethal mutations, conditional mutations

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

7) Describe the mutagenic action of the following two mutagens: 5-bromouracil and ultraviolet light.
Answer: The mutagen 5-bromouracil is an analog of thymine, which anomalously pairs with guanine. Ultraviolet light causes thymine dimers.

8) Considering the electromagnetic spectrum, identify likely mutagens from the following list: radio

waves, microwaves, infrared, ultraviolet, X-rays, gamma rays, cosmic rays.
Answer: ultraviolet, X-rays, gamma rays, cosmic rays

9) What is meant by the term *photoreactivation* repair?
Answer: Photoreactivation repair, discovered in 1949, is a process described in *E. coli* in which UV-induced DNA damage can be partially reversed if cells are briefly exposed to light (in the blue range of the visible spectrum).

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

TRUE or FALSE

11) Loss-of-function mutations eliminate the function of a gene product and may be dominant or recessive.
Answer: TRUE

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.
Answer: TRUE

13) The shorter the wavelength of a radiation source, the greater its likelihood of causing damage.
Answer: TRUE

14) A missense mutation causes premature chain (protein) termination.
Answer: 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
Answer: B

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

Answer: A

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
Answer: C

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.
Answer: B

5) Chronic myelogenous leukemia appears to be associated with a chromosomal rearrangement. How is a chromosomal rearrangement responsible for this disease?
Answer: Joining of chromosomes 9 and 22 through translocation generates a hybrid gene *bcr/c-abl* that produces a protein that is inappropriately active and causes the disease.

6) Which three stages or transitions in the cell cycle seem to serve as points of control (checkpoints)?
Answer: G1/S, G2/M, M

7) Describe the cellular and molecular function of the *ras* gene family and the consequences of mutations in *ras*.
Answer: The *ras* gene family encodes a protein that is involved with signal transduction in the cell membrane. Point mutations may cause changes in function that promote abnormal signaling, thus stimulating uncontrolled cell growth.

8) What is the name of a normal gene that serves to promote cellular division?
Answer: proto-oncogene

9) What are two properties shared by all types of cancer?
Answer: uncontrolled cell replication, metastasis

10) Name three human cancers with a genetic predisposition. What appears to be the genetic cause of each?
Answer: retinoblastoma (RB), mutations in *RB1*; breast cancer: mutations in *BRCA1* and *BRCA2*; FAP-associated colon cancer: mutations in *APC* (and later in *DCC* and *p53*)

11) List at least three environmental agents or factors that are known to cause cancer.
Answer: radiation, a variety of chemicals, sunlight, certain diets, tobacco

TRUE or FALSE

12) Any agent that causes damage to DNA is a potential carcinogen.
Answer: TRUE

13) There are several checkpoints in the mitotic cell cycle. All occur in the S phase.
Answer: FALSE

14) The gene p53 is called the "guardian of the genome" because it corrects mutations in the spindle apparatus before nondisjunction can occur.
Answer: FALSE

15) A *tumor-suppressor gene* normally functions to limit cell replication.
Answer: TRUE