Jorde: Medical Genetics, 5th Edition

Chapter 2: Basic Cell Biology: Structure and Function of Genes and Chromosomes

Multiple Choice

1. Mutation in fibroblast growth factor receptor 3 (FGFR3)
a. Retinoblastoma
b. Achondroplasia
c. Neurofibromatosis type 1
d. Huntington disease
e. Marfan syndrome

Answer: b

Correct Feedback: b. Mutations in the fibroblast growth factor receptor 3 do cause achondroplasia.

Incorrect Feedback: a. Retinoblastoma is caused by mutations in a tumor suppressor on chromosome 13.
c. Neurofibromatosis type one is caused by a mutation of the neurofibromin gene (which may act as a tumor suppressor) on chromosome 17q
d. Huntington disease is caused by a CAG expanded repeat on the distal tip of chromosome 4p.
e. Marfan patients have mutations of the chromosome 15 gene encoding fibrillin, a connective tissue protein.

2. Abnormal binding of gene product to GAPDH (enzyme involved in glycolysis)
a. Retinoblastoma
b. Achondroplasia
c. Neurofibromatosis type 1
d. Huntington disease
e. Marfan syndrome

Answer: d

Correct Feedback: d. This is a characteristic of Huntington disease.

Incorrect Feedback: a. Retinoblastoma is caused by mutations in a tumor suppressor on chromosome 13.
b. Mutations in the fibroblast growth factor receptor 3 cause achondroplasia.
c. Neurofibromatosis type one is caused by a mutation of the neurofibromin gene (which may act as a tumor suppressor) on chromosome 17q
e. Marfan patients have mutations of the chromosome 15 gene encoding fibrillin, a connective tissue protein.

3. phosphorylation of gene product by cyclin-dependent kinases (CDK); binding of gene product to transcription factors such as E2F
a. Retinoblastoma
b. Achondroplasia
c. Neurofibromatosis type 1
d. Huntington disease
e. Marfan syndrome

Answer: a
Correct Feedback: a. The retinoblastoma gene product is phosphorylated by a CDK and then binds to transcription factors.

Incorrect Feedback: b. Mutations in the fibroblast growth factor receptor 3 cause achondroplasia.
c. Neurofibromatosis type one is caused by a mutation of the neurofibromin gene (which may act as a tumor suppressor) on chromosome 17q
d. Huntington disease involves abnormal binding of gene product to GAPDH (enzyme involved in glycolysis)
e. Marfan patients have mutations of the chromosome 15 gene encoding fibrillin, a connective tissue protein.

4. Mutations in fibrillin gene
a. Retinoblastoma
b. Achondroplasia
c. Neurofibromatosis type 1
d. Huntington disease
e. Marfan syndrome

Answer: e

Correct Feedback: e. Marfan patients have mutations of the chromosome 15 gene encoding fibrillin, a connective tissue protein.

Incorrect Feedback: a. The retinoblastoma gene product is phosphorylated by a CDK and then binds to transcription factors.
b. Mutations in the fibroblast growth factor receptor 3 cause achondroplasia.
c. Neurofibromatosis type one is caused by a mutation of the neurofibromin gene (which may act as a tumor suppressor) on chromosome 17q
d. Huntington disease involves abnormal binding of gene product to GAPDH (enzyme involved in glycolysis)

5. Which of the following could produce an XY female?
a. Deletion of the Sry gene
b. Point mutation in the Sry gene
c. Translocation of the Sry gene to the X chromosome during meiosis in the father
d. None of the above

e. All of the above

Answer: e

Correct Feedback: e. All of the above could produce an XY female.

Incorrect Feedback: a. This is true, but it is not the only true answer.
b. This is true, but it is not the only true answer.
c. This is true, but it is not the only true answer.
d. There are true answers.

6. Which of the following is **not** a characteristic of cystic fibrosis?
a. Chloride channel defect
b. Hyperabsorption of intracellular sodium
c. Elevated sweat chloride
d. Fibrous ovarian cysts
e. Pancreatic insufficiency

Answer: d

Correct Feedback: This is not a characteristic of cystic fibrosis.

Incorrect Feedback: This is a characteristic of cystic fibrosis.

7. Each of the following chromosome abnormalities involves a 20 megabase region of the long arm of chromosome 5 (5q). Which abnormality is **most** likely to cause severe disease?
a. Deletion of the region
b. Duplication of the region
c. A balanced translocation involving the region (i.e., in the translocation carrier)
d. Pericentric inversion
e. Paracentric inversion

Answer: a

Correct Feedback: This is the most likely to cause severe disease.

Incorrect Feedback: This can cause problems, but they are not as likely to be as severe as a deletion of the entire gene.

8. Which of the following diseases is a good example of locus heterogeneity?
a. Prader-Willi syndrome
b. Myotonic dystrophy
c. Osteogenesis imperfecta
d. Duchenne muscular dystrophy
e. Hemophilia A

Answer: c

Correct Feedback: c. Locus heterogeneity is where genes have more than on discernible effect. OI effects bones, teeth, and sclera.

Incorrect Feedback: a. Prader-Willi syndrome is a good example of genomic imprinting.
b. Myotonic dystrophy is a good example of anticipation.
d. Duchenne muscular dystrophy is an X-linked disease.
e. Hemophilia A is an X-linked disease.

9. Why are some autosomal dominant disorders (e.g., Marfan syndrome) seen more commonly in the offspring of older fathers?
a. Replication errors accumulate as sperm-producing stem cells continue to divide
b. Rate of nondisjunction increases in older males
c. Recombination rates increase in older males
d. All spermatocytes are produced during male embryonic development, so older males produce older sperm cells
e. None of the above

Answer: a

Correct Feedback: a. This is why some autosomal dominant disorders are seen more commonly in the offspring of older fathers.

Incorrect Feedback: b. This is seen in older mothers.
c. This is not true.
d. This is not true.
e. There is a correct answer

10. A woman with phenotypically normal parents has two brothers with Duchenne muscular dystrophy. She experiences mild muscle weakness in her legs. Which of the following mechanisms is **most likely** to be directly involved?
a. Germline mosaicism
b. Skewed X inactivation
c. Mutation near the pseudoautosomal region of the Y chromosome
d. New mutation in this woman
e. Nondisjunction of her mother's X chromosomes

Answer: b

Correct Feedback: b. This is most likely to be directly involved.

Incorrect Feedback: a. This wouldn't explain why a heterotroph would manifest the disease.

c. A female would not have a Y chromosome.
d. If this were the case it would be unlikely that her brothers had the disease.
e. Duchenne muscular dystrophy is not a chromosomal disease.

11. Consider a fetus affected with one of the following conditions. For which condition is spontaneous loss during pregnancy most likely?
a. Down syndrome
b. Neurofibromatosis type 1
c. Retinoblastoma
d. Huntington disease
e. Trisomy 18

Answer: e

Correct Feedback: e. Chromosomal abnormalities are the number one cause of fetal loss, and trisomy 18 is less compatable with survival than trisomy 21.

Incorrect Feedback: a. Chromosomal abnormalities are the number one cause of fetal loss, but trisomy 18 is less compatable with survival than trisomy 21
b. Chromosomal abnormalities are the number one cause of fetal loss.
c. Chromosomal abnormalities are the number one cause of fetal loss.
d. Chromosomal abnormalities are the number one cause of fetal loss.