Genetic Disorders

Name: _____ **Date:** _____

Students must provide an explanation for all problems. Students must have parent signature prior to submission.

1. A human hereditary disorder that may result in mental retardation is 1. _____

   A. phenylketonuria
   B. hemophilia
   C. sickle-cell anemia
   D. albinism

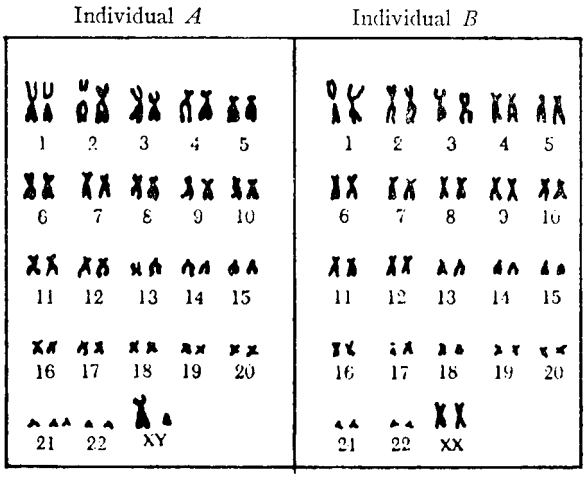
2. In which hereditary disease do the abnormal hemoglobin molecules differ from normal hemoglobin molecules by only a single amino acid? 2. _____

   A. hemophilia
   B. albinism
   C. phenylketonuria
   D. sickle-cell anemia

3. Which statement best describes amniocentesis? 3. _____

   A. Blood cells of an adult are checked for fragility.
   B. Saliva of a child is analyzed for amino acids.
   C. Urine of a newborn is analyzed for the amino acid phenylalanine.
   D. Fluid surrounding an embryo is removed for cellular analysis.

4. The charts show human chromosomes arranged in pairs. 4. _____



The chromosome numbered 1 through 22 are known as

A. ribosomes B. lysosomes C. centrosomes D. autosomes

5. The preparation of these charts for individuals *A* and *B* is known as 5. _____

A. microsurgery B. karyotyping

C. blood typing D. chemical screening

6. Which genetic disorder in individual *A* is indicated by the number of chromosomes labeled 21? 6. _____

A. phenylketonuria (PKU) B. Tay-Sachs

C. sickle-cell anemia D. Down's syndrome

7. Techniques for Detecting Genetic Disorders

(1) Karyotyping (3) Blood Analysis

(2) Urinalysis (4) Amniocentesis

PKU is a disorder that results from the inability to synthesize a single enzyme necessary for metabolism of phenylalanine. It is most easily detected in newborns by

A. 1 B. 2 C. 3 D. 4

7. _____

8. Down syndrome is a genetic disorder caused by the presence of an extra chromosome number 21 in the body cells of humans. This extra chromosome most likely is a result of

A. mitotic cell division in the brain

B. development of an unfertilized egg

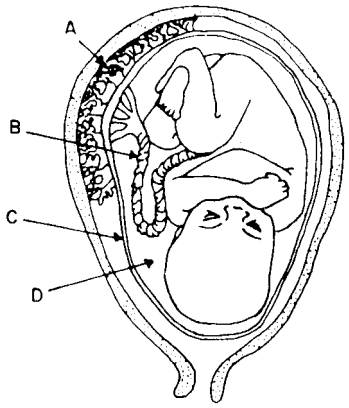
C. multiple allelic pairs of genes

D. nondisjunction during meiosis

8. _____

9. From which area would fluid be removed to detect genetic disorders by amniocentesis?

A. *A* B. *B* C. *C* D. *D*



9. _____

10. Select the genetic disorder, *chosen from the list below*, that is best described by the statement shown. 10. _____

A disorder characterized by the formation of abnormally shaped red blood cells

A. Phenylketonuria PKU
B. Sickle-cell anemia
C. Tay-Sachs disease
D. Hemophilia

11. X rays, formaldehyde and asbestos fibers are all similar in that they are 11. _____

A. animal preservatives
B. used to treat diseases
C. used to diagnose diseases
D. motogenic agents

12. Select the genetic technique, *chosen from the list below*, that is best described by the statement shown. 12. _____

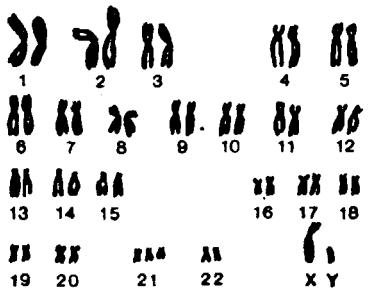
The presence or absence of a particular enzyme is determined by chemical analysis of a blood sample.

A. Screening
B. Amniocentesis
C. Cloning
D. Genetic engineering

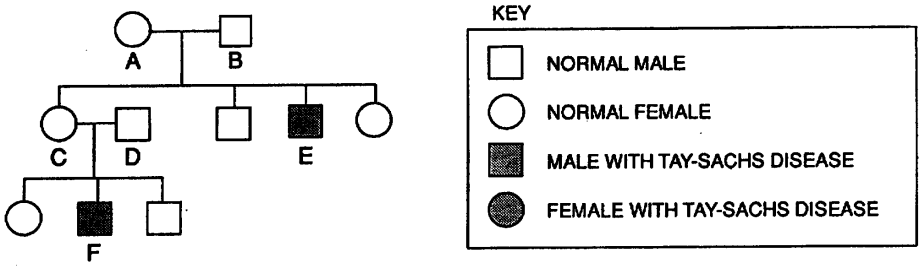
13. The individual from whom these chromosomes were taken is a 13. _____

A. male B. female

C. hermaphrodite D. polyploid



14. The chart represents the inheritance of Tay-Sachs disease in a family. 14. _____



Answer the following question(s) based on the chart shown and on your knowledge of biology.

What are the genotypes of individuals *A* and *B* with regard to Tay-Sachs disease?

A. One must be homozygous dominant and the other must be homozygous recessive.

B. One must be homozygous dominant and the other must be heterozygous.

C. Both must be homozygous.

D. Both must be heterozygous.

15. Base your answer(s) to the following question(s) on the passage below and on your knowledge of biology. 15. _____

Ibuprofen Helps Patients with Cystic Fibrosis

A faulty version of the CFTR gene causes the disease cystic fibrosis (CF). This gene is found in 1 in 25 Caucasians in the United States. A person who inherits a copy of this gene from each parent develops CF. Thick mucus builds up in the lungs of CF patients, leaving them vulnerable to infections. Over time, this repeated cycle of illness and inflammation causes structural damage to the lungs of the patient.

In a recent study, the common pain reliever ibuprofen significantly reduced lung damage caused by cystic fibrosis. This study included 85 CF patients between the ages of 5 and 39. Half of those patricipating in the study were given a tablet containing ibuprofen, and the other half were given a placebo (a tablet containing no ibuprofen). Ibuprofen, and along with other treatments, most benefited CF patients between the ages of 5 and 13. Patients taking ibuprofen suffered less inflammation of the bronchial tubes. Lung deterioration in the children taking ibuprofen was nearly 90% slower than expected. Among those patients taking ibuprofen, lung capacity declined by only 2%, while those taking the placebo experienced a decline of 16%.

Researchers recommended that doctors begin the new therapy with their cystic fibrosis patients. However, the treatment involves taking large doses of ibuprofen, which can cause serious side effects, including stomach and kidney damage. The researchers warn people with cystic fibrosis no to take ibuprofen without talking with their doctors first.

Thirty years ago, most CF patients died before the age of 5. Today, many CF patients live into their 30's. A new drug for CF, DNase, was approved in 1994. Trials are also being done using gene therapy to correct the faulty gene found in cystic fibrosis patients. Since ibuprofen therapy delays the progression of the disease, it is hoped that more patients will be able to benefit from gene therapy when it becomes available for general use.

Cystic fibrosis results when an individual is

A. homozygous for the faulty CFTR gene

B. heterozygous for the faulty CFTR gene

C. given an overdose of ibuprofen

D. exposed to a person with this disease