Name _____ Class _____ Date _____

14.2 Human Genetic Disorders

From Molecule to Phenotype

1. The boxes below each show a step to explain how genetic disorders have a molecular basis. Number them so that the steps are in the correct order.

A change in phenotype results. _____	A gene's DNA sequence changes. _____	The amino acid sequence that alters a protein changes. _____

For Questions 2–7, write the letter of the correct answer on the line at the left.

_____ **2.** How many human genetic disorders are known?

A. three

B. about 20

C. about 100

D. thousands

_____ **3.** The inherited disease in which hemoglobin molecules clump into long fibers, changing the shape of blood cells is

A. cystic fibrosis.

B. sickle cell disease.

C. Huntington's disease.

D. Klinefelter's syndrome.

_____ **4.** What happens to the CFTR gene in individuals who have cystic fibrosis?

A. The entire gene is deleted.

B. The entire gene is duplicated.

C. Three bases are deleted, causing one amino acid to be missing.

D. Three bases are duplicated, causing one amino acid show up about 40 times.

_____ **5.** Why are individuals who are heterozygous for the cystic fibrosis allele unaffected by the disease?

A. They have an extra copy of the allele on their X chromosome.

B. Cystic fibrosis only occurs in males, so females are unaffected.

C. They make enough of a particular protein to allow their cells to work properly.

D. Their cells can transport chloride ions through diffusion channels.

_____ **6.** How might the allele that causes a disease stay in the population if it is fatal to those who have the disease?

A. It is present only in heterozygotes.

B. It makes the heterozygote resistant to a fatal disease.

C. It disappears but is continuously replaced by mutations.

D. It occurs only in certain geographic areas.

_____ **7.** What advantage do individuals with one sickle cell allele have?

A. a stronger resistance to malaria

B. immunity to typhoid fever

C. more rigid red blood cells

D. no advantage

Chromosomal Disorders

9. What is trisomy?

10. What happens when a male has XXY sex chromosomes?

Apply the Big idea

11. Most of the genetic disorders you have learned about are the result of a change in DNA sequence, as with cystic fibrosis, or the presence of an extra chromosome, as with Down syndrome. The exception is Turner's syndrome. Women with Turner's syndrome have only 45 chromosomes. They are missing an X chromosome. This disorder is the *only* case in which a person can survive with one less chromosome. What does this tell you about how genetic information is inherited in humans?