



## Genetics Home Reference

Your Guide to Understanding Genetic Conditions

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### Explore Genetics Home Reference

Learn about the main features of the Genetics Home Reference web site (<http://ghr.nlm.nih.gov>) by answering the following questions:

1. Alström syndrome is caused by mutations in what gene? ANSWER: \_\_\_\_\_
2. What genetic condition is caused by mutations in the MT-ND4L gene?  
ANSWER: \_\_\_\_\_
3. What structure in the cell helps process molecules and transports them to specific destinations inside or outside the cell? (You might find the first chapter of the Handbook helpful in answering this question.) ANSWER: \_\_\_\_\_
4. The DMD gene is located on which chromosome? ANSWER: chromosome \_\_\_\_\_
5. What is the inheritance pattern for cystic fibrosis? ANSWER: \_\_\_\_\_
6. According to information in “About,” who sponsors the Genetics Home Reference?  
ANSWER: The U.S. \_\_\_\_\_
7. Look up the glossary definition for the term “protein product.” The definition comes from the Human Genome Project Information, which is part of what government agency? ANSWER: U.S. \_\_\_\_\_
8. Which of the following is listed under “Research Resources” for the TP53 gene: GeneTests, Entrez Gene, or PubMed? ANSWER: \_\_\_\_\_
9. Find the summary for chromosome 13. What disorder is caused by a deletion in a region of chromosome 13 known as 13q14? ANSWER: \_\_\_\_\_

Fill in the blanks below using the first letter from each of your answers.  
What genetic condition is spelled out?

\_\_\_\_\_ DISEASE