## Period\_

## **Chromosomal Disorders Webquest**

There are many inherited disorders in the human population. This webquest covers some of these disorders. Using the websites listed below, answer the following questions.

Use the following website <u>http://www.ygyh.org/cf/whatisit.htm</u>) to answer the questions below.

- 1. What are some of the major symptoms of cystic fibrosis?
- 2. In what ethnic group(s) is CF most common?
- 3. How is CF inherited?
- 4. What type of screening is done for CF?
- 5. What treatments are available to CF patients?

Use <u>http://www.ygyh.org/dmd/whatisit.htm</u> to answer the questions that follow about Duchenne Muscular Dystrophy.

- 6. How is DMD inherited?
- 7. What are the symptoms of DMD?
- 8. How is DMD diagnosed?
- 9. How is DMD treated?

Use <u>http://www.ygyh.org/fragx/whatisit.htm</u> to answer the following questions.

- 10. Where does the name Fragile X Syndrome come from?
- 11. What are the symptoms of this disorder?
- 12. What type of screening is done for Fragile X?
- 13. What treatments are available for people with Fragile X Syndrome?

Use the website, <u>http://www.nlm.nih.gov/medlineplus/klinefelterssyndrome.html</u> to answer questions about Klinefelter's syndrome.

14. How is Klinefelter's inherited?

15. What are some of the symptoms of Klinefelter's?

16. How is screening for Klinfelter's done?

17. What are some of the treatments available to males with Klinefelter's?

Use the website <u>http://www.medhelp.org/lib/pku.htm</u> to answer the following questions about Phenylketonuria (PKU).

18. What causes PKU?

- 19. What is the mode of inheritance of PKU?
- 20. How are infants tested for PKU?
- 21. How is PKU treated?

Use the following website <u>http://www.pediatriconcall.com/forpatients/CommonChild/turner\_syndrome.asp</u> to answer questions about Turner's Syndrome

- 22. How is Turner's Syndrome inherited?
- 23. How is Turner's Syndrome diagnosed?
- 24. What are some of the characteristics and symptoms of Turner's Syndrome?
- 25. What are some of the treatments for Turner's Syndrome?

Use <u>http://www.achondroplasia.co.uk/</u> to answer the following questions. 26. What is the common name of Achondroplasia?

- 27. How is Achondroplasia inherited?
- 28. What are some of the characteristics of individuals with Achondroplasia?

Use the following website <u>http://www.pwsausa.org/faq.htm</u> to answer questions about Prader-Willi Syndrome 29. What are some of the characteristics/symptoms of Prader-Willi Syndrome?

30. What parent donates the defective chromosome that causes this disorder?

31. Which chromosome is associated with this disorder?

32. What is wrong with that particular chromosome?

Use <u>http://www.nlm.nih.gov/medlineplus/ency/article/001660.htm</u> and the yahoo site to answer the questions below concerning Trisomy 13.

33. What is the other name for Trisomy 13?

- 34. What does trisomy mean?
- 35. What are some of the symptoms of this disorder?
- 36. What is the life expectancy of individuals with this disorder?

<u>http://americanpregnancy.org/birth-defects/down-syndrome/</u> to answer the following questions about Trisomy 21. 37. What is the common name of Trisomy 21?

38. What are some of the features that make Trisomy 21 easy to recognize?

39. What are the 3 genetic variations of the disorder?

## **Conclusions page:**

Scientists working around the world have mapped the 46 human chromosomes. The project is called the Human Genome Project. Today, these mapped chromosomes with their genes can be studied to determine what genes may cause different characteristics or disorders in humans.

One such mapping site is the National Center for Biotechnology's internet site, "The Human Gene Map." <u>http://www.ncbi.nlm.nih.gov/books/NBK22266/</u>

- **1.** Use this website to locate the chromosome and gene responsible for <u>4</u> different human genetic disorders.
- **2.** Click on different chromosomes. Then click on the disease located on the chromosome you are interested in.
- **3.** For each include the name of the disease, chromosome it is assosciated with, brief description, and if it can be inherited or not.

1.\_\_\_\_Chromosome

2.\_\_\_\_Chromosome

3.\_\_\_\_Chromosome

4.\_\_\_\_Chromosome

- 1. Please provide a short summary of what you did during this lab and what you learned.
- 2. What is the difference between a chromosomal mutation and a gene mutation?
- 3. How do changes in DNA affect proteins?

4. How can the "Human Genome Project" help our understanding of diseases?