Biology: Genetics

**Lab: How Are Genetic Disorders Identified in Humans?**

**Introduction:** Several genetic disorders in humans are caused by additional, missing, or damaged chromosomes. One way to study genetic disorders is to observe the chromosomes themselves. Cells from people with disorders are grown in a laboratory, stained and set on glass slides. The slides are observed under a microscope, counted and checked for abnormalities. Then a photograph of the chromosomes is enlarged and chromosomes are individually cut out and arranged in matching pairs. This is called a **karyotype.**

In this lab, you will study normal karyotypes and make a karyotype to study for any abnormalities. There will be a variety of karyotypes around the room. Do not be concerned if you are not getting the same result as the person next to you.

**Materials**: scissors tape chromosome smear

**Procedure**: Study figures 5.2 and 5.3 as examples of normal sets of chromosomes. Note that the last pair of chromosomes may not look alike while the other pairs do. The normal human cell has 46 chromosomes.

• You will use the page titled Karyotype Placement Grid to compile your data.

• Record the number of the smear you are given at the top of the grid.

• Your teacher will give you a human chromosome smear.

• Study this page of chromosomes looking closely at size and patterns.

• Cut out the chromosomes being careful not to misplace any. Remember many disorders involve missing or additional chromosomes.

• Line up the chromosomes on the grid and check your results before you tape them into order.

• Carefully study your results.

Information on chromosome disorders is included in this lab assignment and you will find additional information in your textbook, other books, or on the Internet.

**Clean Up**: Put all scraps of paper in the wastebasket. The scissors and tape should be put back in the bins where you got them.

**Note:** Attached are one grid and the 2-page disorder information sheet. The numbered chromosome smear page will be handed out separately.

**Analysis**

**1.** What, if any irregularities do you see in the grid you made?\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**2.** The number of the smear you used was \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**3.** What pair of chromosomes is abnormal? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**4.** What is the name of the disorder this arrangement would cause? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**5.** Describe the results of this disorder \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

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**6.** What relationship exists between the number of a chromosome and its size? \_\_\_\_\_\_\_

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**7.** Identify the sex of the person whose smear you studied. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**8.** For each of the disorders listed here, identify what mutation or abnormality would cause the disorder:

Downs Syndrome - \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Philadelphia translocation - \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Edwards syndrome - \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

Patau syndrome - \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

**9.** This karyotype was made from a 17 year-old person. Will it look any different when the person is 45 years old?

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**Information on chromosome Disorders**

*Adapted from “Karyotype Success Rate Increases With Stylized Chromosomes” by Caroline Purser*, The American Biology Teacher, *September 1987.*

**A. Cri-Du-Chat**

This disorder is the result of a deletion in the upper arm of one chromosome number 5. Babies have a cry which sounds like that of a cat in distress (hence the name, which means cry of the cat). This is the result of an improperly developed larynx. Cri-du-chat babies are severely mentally retarded and have a small cranium. The incidence of this syndrome is 1 in 100,000 live births.

Karyotype: 46XX or 46XY with a deletion in chromosome #5

**B: Down Syndrome**

This syndrome is the result of an extra chromosome number 21. The disorder results in individuals who are short in stature and have broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult, and mental retardation to varying degrees. These individuals are prone to respiratory infections, heart defects, and leukemia. The risk of having a baby with Down Syndrome increases with the age of the mother, ranging from 1 in every 1,500 live births for mothers in their early twenties to 1 in every 70 for mothers over 35 and 1 in every 25 for mothers 45 or older.

Karyotype: 47XX or 47XY with 3 of chromosome #21

**C. Philadelphia Translocation**

This disorder is the result of part of one chromosome number 9 being inserted or translocated into chromosome number 22. Individuals with this disorder suffer from chronic myelogenous leukemia believed to be the result of two genes normally not on the same chromosome being expressed on the same chromosome and producing an unusual protein which causes the disease.

Karyotype: 46XX or 46XY with a chromosome #9 translocation to chromosome #22

**D. Turner Syndrome**

This condition is the result of the lack of a second sex chromosome; the resulting individual has one X and no second X or Y chromosome. These girls tend to be short and have a stocky build but appear to be normal before puberty. At puberty, no secondary sexual characteristics develop. They produce no eggs, and menstruation and breast development do not occur. The frequency is 1 in 2,500 live female births.

Karyotype: 45X with 1 X chromosome

**E. Klinefelter Syndrome**

This disorder is the result of having two X chromosomes and one Y chromosome. Individuals with this syndrome appear to be normal males, but have characteristics, which include tall stature, small testicles, and sterility. This condition occurs once in every 1,000 live male births.

Karyotype: 47XXY

**F. Jacobs**

This condition is the result of an extra Y chromosome. Men with the condition are tall and have low mental ability. This condition occurs at a rate of 1 in every 1,000 live male births.

Karyotype: 47 XYY

**G. Triple X**

Individuals with this condition have three X chromosomes. However, no specific physical or mental abnormalities are associated with an extra X chromosome, and women who have this are normal and fertile.

Karyotype: 47 XXX

**H. Edwards Syndrome**

This syndrome is the result of having an extra chromosome number 18. Symptoms of this condition include severe retardation and physical malformations such as an elongated skull, a very narrow pelvis, low set ears, and very small mouth and teeth. Nearly all babies born with this condition die in early infancy. The frequency of occurrence is 1 in every 5,000 live births.

Karyotype: 47 XX or 47 XY with 3 of chromosome #18

**I. Patau Syndrome**

This condition is the result of an extra chromosome number 13. This results in severely abnormal cerebral functions and almost always leads to death in early infancy. A baby with this syndrome has a very pronounced cleft lip and palate, a small cranium, and nonfunctional eyes as well as heart defects and mental retardation. The frequency of this occurrence is 1 in every 15,000 live births.

Karyotype: 47 XX or 47 XY with 3 of chromosome #13