

KARYOTYPING LAB

Introduction:

Scientists have developed several different tools and techniques for studying chromosomes, genes and base pairs in humans and other organisms. One of the most useful techniques is karyotyping. **A karyotype is a photograph of all of an organism's chromosomes.** The chromosomes in the karyotype are arranged in homologous pairs according to size (largest to smallest). Homologous pairs can be determined by centromere placement, equal length of top and bottom arms as well as similar band placement on each arm. *Karyotyping helps doctors diagnose and treat genetic disorders.* Doctors use a normal human karyotype and compare it to the karyotype of a patient to determine if there are abnormalities. Some of the characteristics the physician will compare are:

- A. Total number of chromosomes - normal humans have 46 chromosomes (23pairs), so if the number is higher or lower then an abnormality exists
- B. Homologous pairs for the first 22 pairs of chromosomes - once centromeres are aligned, top and bottom arms are of equal length and if not then an abnormality exists
- C. Sex Chromosomes (23rd pair) - if female, then 2 homologous X chromosomes (XX) will be present and if male, an X chromosome and a Y chromosome (XY) will be present, so if there are additional or fewer sex chromosomes then an abnormality exists. The X chromosome is much larger than the Y.

Procedure:

1. From the chromosome scatter sheet you received, carefully cut out each of the chromosomes into a rectangles, so they will fit into the layout worksheet.
2. Arrange the chromosomes into homologous pairs, using your Karyotype Reference Sheet as a guide. Hint: Laying your cut chromosomes directly on top of the matching chromosomes on the Karyotype Reference Sheet will help you in this process.
3. Once all chromosomes are laid down on the Reference Sheet begin gluing or taping each pair onto the Karyotype Layout Worksheet. Hint: You may have abnormalities in your karyotype (more or less than 46 chromosomes), so maintain the number of chromosomes you were given.
4. Upon completion of gluing or taping all chromosomes to the Layout Worksheet, answer the Discussion Questions using the background information, your karyotype and Explanation of Chromosome Disorders

Discussion Questions:

Write the answers to these questions on the back of the Karyotype Layout Worksheet.

Use Background Information to answer the following questions.

1. Define Karyotype.
2. What can karyotypes be used to determine?
3. How many chromosomes do normal humans have?
4. What does the 23rd pair of chromosomes determine?
5. The first 22 pairs of chromosomes are homologous in a normal person. Is that "always true for the 23rd pair of chromosomes? Explain your answer.

Use your karyotype to answer the following questions.

6. What is the gender (sex) of your baby?
7. If the karyotype was abnormal, what is the disorder your baby has (use the Explanation of Chromosome Disorders Handout)?
8. Describe the disorder your baby has. If your baby is normal write "Normal Baby".

Use the Explanation of Chromosome Disorders Handout to answer the following questions.

9. What pair of chromosomes does Patau Syndrome affect?
10. What are 2 characteristics of Edward's Syndrome?
11. What are 2 characteristics of Down Syndrome?
12. Looking at the 8 explanations, what is one problem found in most of the disorders?
13. What are two disorders that only affect males?
14. What are two disorders that only affect females?
15. Of the 8 disorders discussed, which is the only one that is caused by a missing piece of a chromosome, not an entire missing or extra chromosome?

EXPLANATION OF CHROMOSOME DISORDERS

Cri-du-chat

Babies with the "cry of the cat" syndrome have a cry which sounds like that of a cat in distress because the infant's larynx is improperly developed. The cause of this condition is a deletion of about half of the short arm of chromosome number five. Cri-du-chat babies are severely mentally retarded, have a small cranium, a small jaw and a moon-shaped face. The incidence of this syndrome is 1/100,000 live births.

Karyotype: 46XX or 46XY with one chromosome #5 upper arm deletion

Down Syndrome

Trisomy 21, one of the most common causes of mental retardation is due to an extra chromosome 21. This results in a number of characteristic features, such as short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult and mental retardation. Individuals with this syndrome have a high incidence of respiratory infections, heart defects and leukemia. The average risk of having a child with trisomy 21 is 1/750 live births. Mothers in their early twenties have a risk of 1/1,500 and women over 35 have a risk factor of 1/70, which jumps to 1/25 for women 45 or older.

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

Edward's Syndrome

This syndrome, trisomy 18 (an extra chromosome 18), produces severe mental retardation and a highly characteristic pattern of malformations such as elongated skull, a very narrow pelvis, rocker bottom feet, malformed heart and a grasping of the two central fingers by the thumb and little finger. In addition, the ears are often low set and the mouth and teeth are small. Nearly all babies born with this condition die in early infancy. The frequency of this syndrome is 1/5,000 live births.

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

Jacobs

A chromosome abnormality which is caused by nondisjunction (non-separation) of the Y chromosome during the second phase of meiosis. This condition only occurs in men and happens when they have an extra Y chromosome. Men with this abnormality are tall, develop heavy cases of acne and have low mental ability. Occurrence is 1/1,000 live male births.

Karyotype: 47XYY (extra sex chromosome - extra Y)

Klinefelter Syndrome

A condition occurring in 1/1,000 male live births. Characteristics associated with this condition are tall stature, small testicles, developed breasts, sterility and mental deficiency. Most men with this syndrome appear normal in other ways. This syndrome only occurs in men and affects the sex chromosomes.

Karyotype: 47XXY or 47XXX (extra sex chromosomes)

Patau Syndrome

This syndrome (trisomy 13 - extra chromosome 13) causes severely abnormal cerebral functions and virtually leads to death in early infancy. The baby has very pronounced clefts of the lip and palate, broad nose, polydactyly (extra fingers and toes), small cranium and nonfunctional eyes. Heart defects and severe mental retardation are also part of the clinical picture. The frequency is 1/15,000 live births.

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

Triple X

Superfemale (XXX). Occurs at a frequency of 1/1,000 female live births. No specific abnormalities are associated with this condition. The vast majority of women who have this condition are normal mentally, but may have underdeveloped genitalia and limited fertility. They also may have neuromotor delays. This condition only occurs in females and affects the sex chromosomes.

Karyotype: 47XXX (affects sex chromosomes - extra X chromosome)

Turner Syndrome

This condition happens when an X-carrying sperm fertilizes an ovum that lacks an X, or when a sperm lacking an X or Y chromosome fertilizes an X-bearing egg. This syndrome only affects girls and causes them to be missing an X chromosome (XC). These girls appear to be normal before puberty, although they are shorter and have a chunky build. At birth, the distinguishable characteristics include a webbed neck. At sexual maturity, the secondary sex characteristics are not developed. There also is no menstruation or breast development and they are usually sterile. The frequency is 1/2,500 live female births.

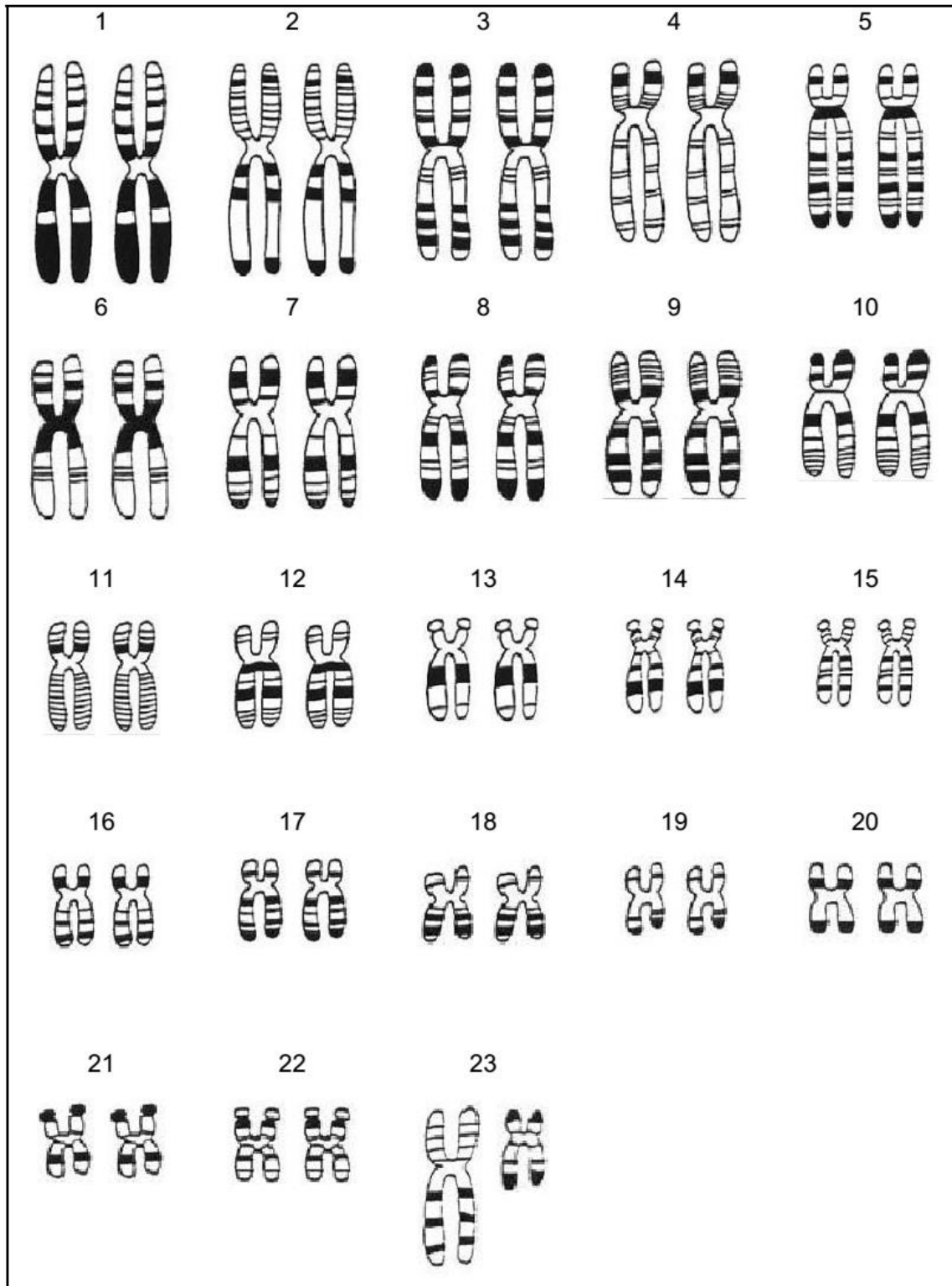
Karyotype: 45X or 45XO (affects sex chromosomes - missing an X)

KARYOTYPE LAYOUT WORKSHEET

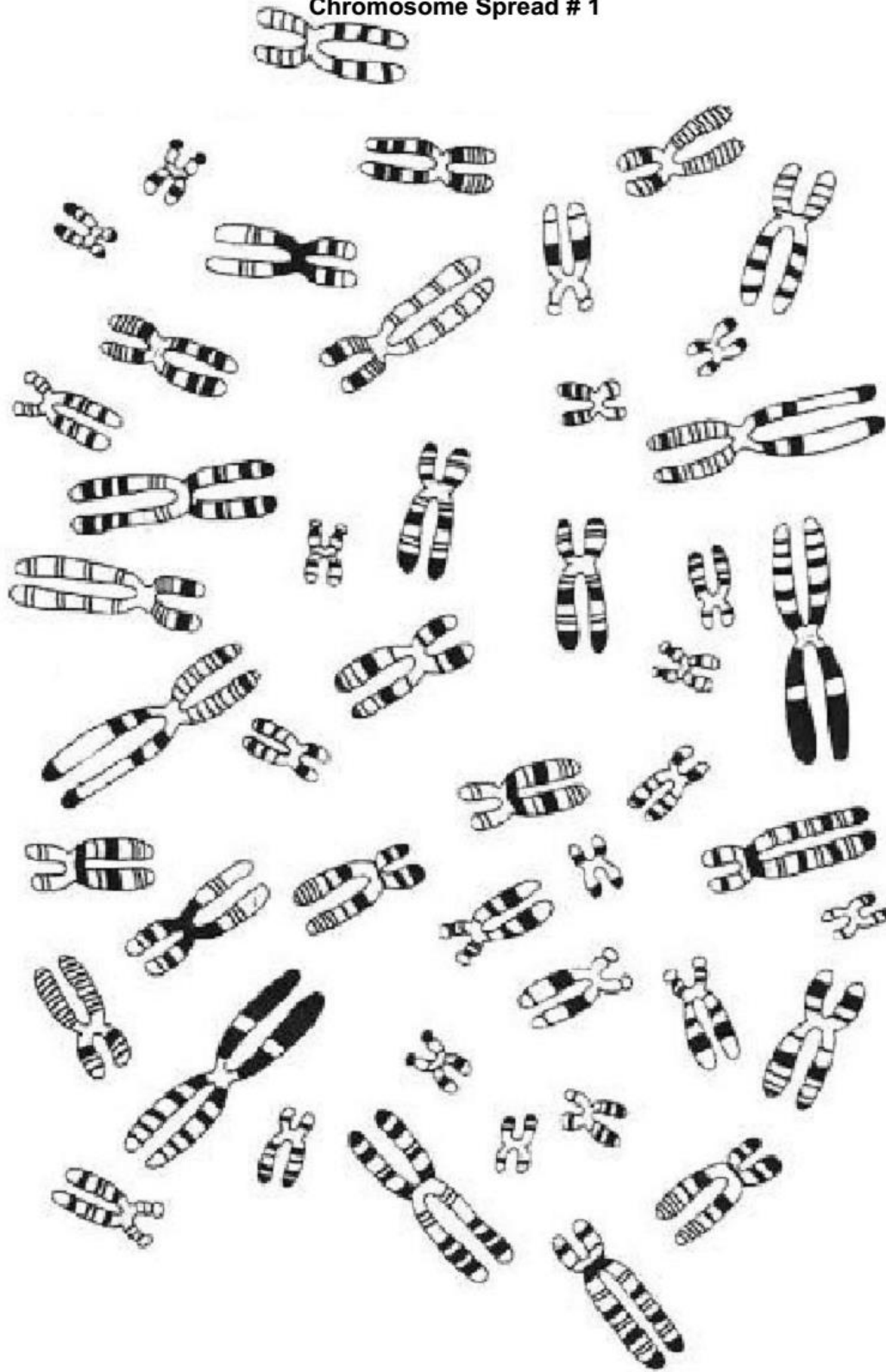
Scatter Sheet Number: _____

1	2	3	4	5	6
7	8	9	10	11	12
13	14	15	16	17	18
19	20	21	22	23 XX or XY	

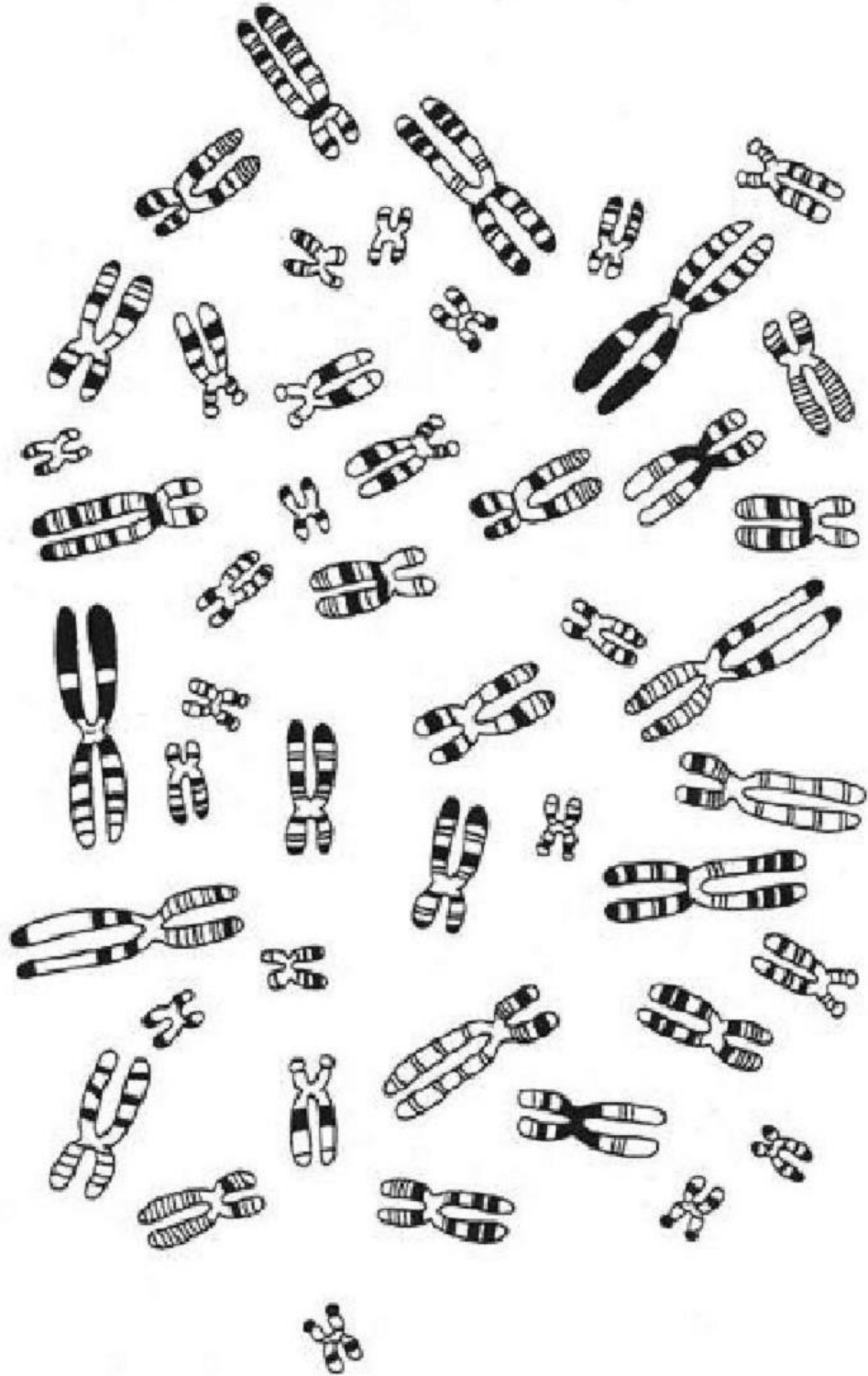
Normal Human Karyotype



Chromosome Spread # 1



Chromosome Spread # 2



Chromosome Spread # 3

