

HUMAN KARYOTYPES AND CHROMOSOMAL DISORDERS

Purpose: Students will cut out prepared karyotypes, arrange chromosomes in the proper place to determine the sex of the person, and if the person has a chromosomal disease.

Materials:

~~Scissors: to cut out the chromosomes~~

~~Tape/glue: used to secure the chromosomes on the key~~

Chromosomes/Chromosomal Disease Guide: the lab centers around these items.

Class set

Procedures:

1. Obtain materials from your teacher

omit → 2. Cut out chromosomes one at a time and secure the chromosomes in the proper place on the karyotype key. Work with one sheet at a time to avoid confusion.

3. When all cutting and taping is complete, analyze karyotype key sheets and identify the sex of the individual and the chromosomal disease.

4. Write a brief description of the chromosomal disease and a rationale ~~on the back of each karyotype key.~~

Chromosomal Disease Guide

Down Syndrome: Down Syndrome is one of the most common form of mental retardation in the population (1 in every 700 births). The disorder is due to an extra 21st chromosome.

People with Down syndrome exhibit the following physical and mental traits. All affected people are short, have an underdeveloped brain and mental retardation (IQ about 50), have heart defects, are susceptible to infections and leukemia, have a single crease along the palm of their hands, simple fingerprints, and their ears are underdeveloped. The infant mortality rate is high due to the heart defects and survival past 40 is rare.

A person with Down syndrome will be able to do the things that a normal 6-7 year old can. Down people can be trained to their full potential and survive outside of institutions. Down Syndrome affects all races and both sexes.

Fragile X Syndrome: This disorder appears mostly in males. People who have this condition are mentally retarded, short, and have very large heads with long and narrow faces. Some people have large and prominent ears. If someone does not display these characteristics, the fragile X syndrome cannot be eliminated as a cause of the mental retardation.

The karyotype shows that people with fragile X syndrome appear to have a broken X chromosome. Females can carry the chromosome but will appear normal due to their second X chromosome.

Fragile X syndrome is the second leading cause (1 in 1,000 to 2,000 males) of mental retardation in newborns in the United States. There is no specific treatment for the disease.

Klinefelters Syndrome: This syndrome occurs in 1 out of 1,000 males. This chromosomal disorder is caused by an extra X chromosome in males. (XXY)

Klinefelters is first discovered in males at puberty. The affected males tend to be taller than average, lanky

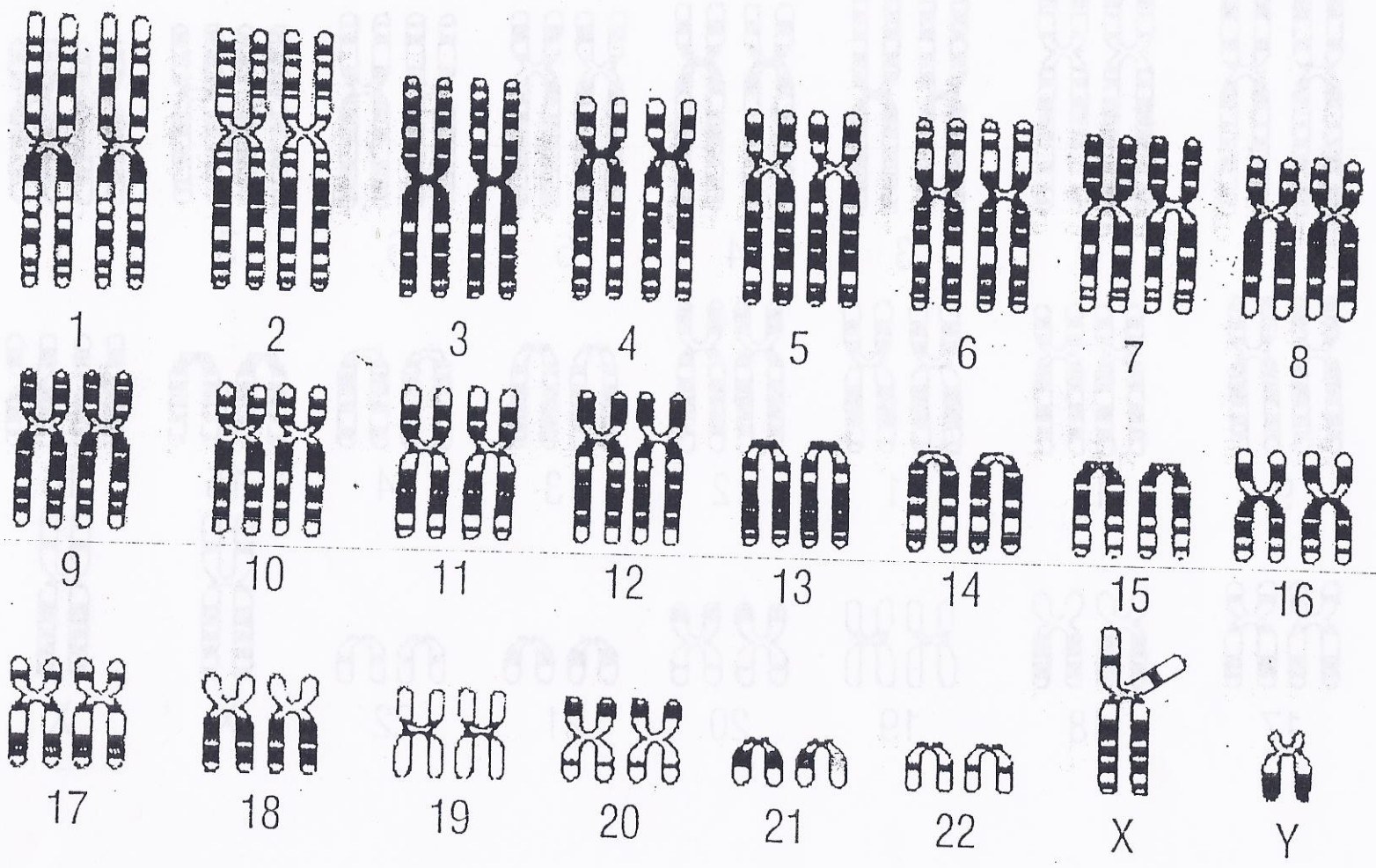
and tend to have feminine muscle development. In fact, there may be some breast enlargement. The testicles in these males do not fully develop causing the affected males to be sterile.

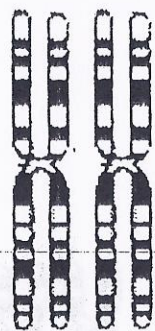
Edwards Syndrome: Edwards syndrome occurs when there are three #18 chromosomes instead of two. About 1 in 8,000 babies are born with Edwards syndrome; however, most of these children die within the first two months of life.

People with Edwards syndrome are born with the following characteristics: mental retardation, receding jaw, low set ears, clenched fists, and heart abnormalities.

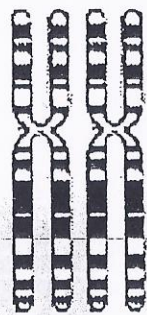
Crit du chat: Crit du chat in French means "cry of the cat". People born with crit du chat often have a cat-like cry.

This genetic disorder can be traced to chromosome #5. The upper arm of one of the chromosome pairs is missing. Crit du chat victims are mentally retarded, have small heads, wide spread eyes, low set chins, small mouths and chins, and low birth weights.

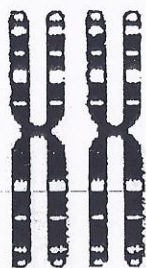




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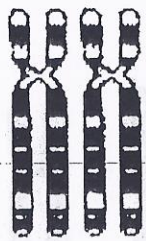
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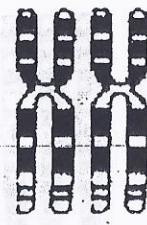
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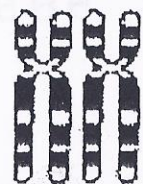
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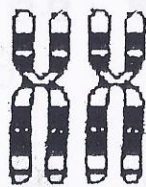
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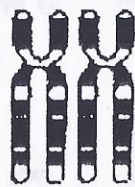
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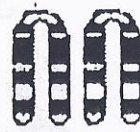
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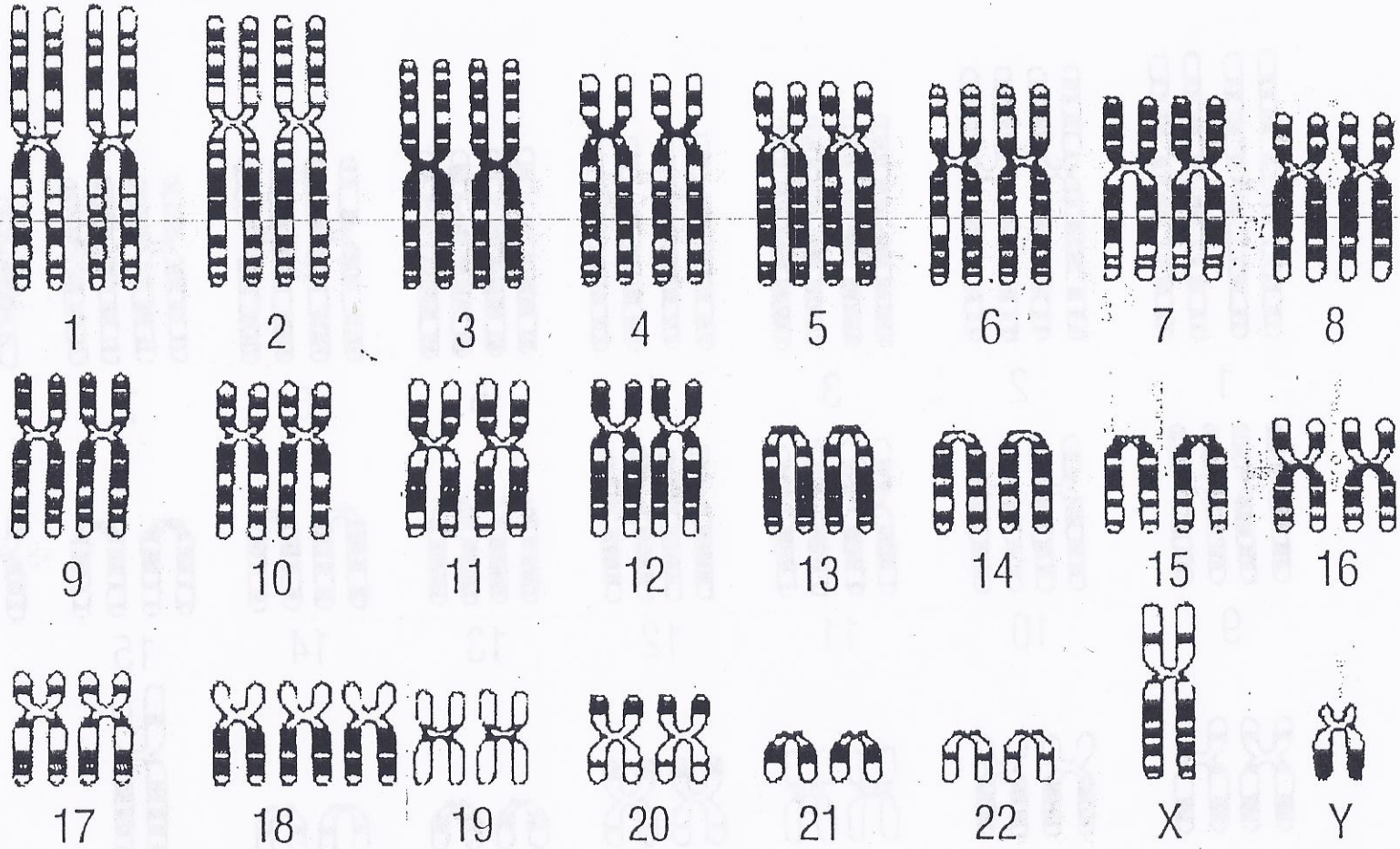
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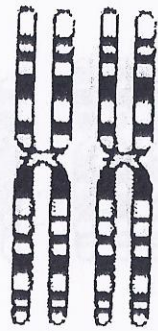


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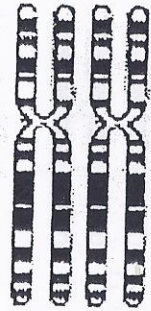


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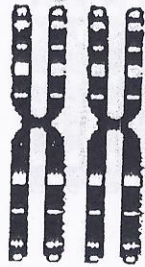




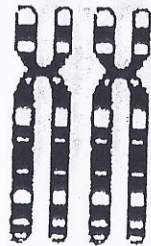
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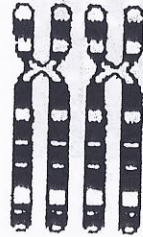
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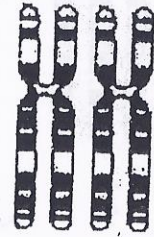
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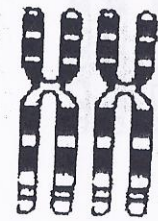
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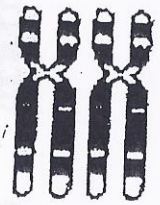
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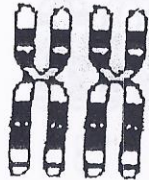
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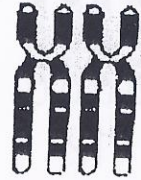
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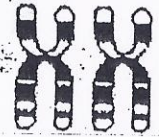
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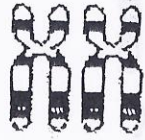
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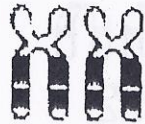
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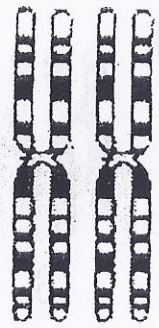
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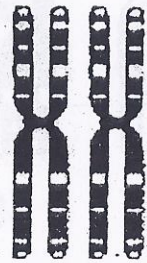
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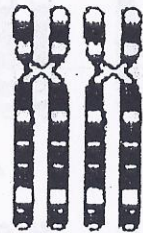
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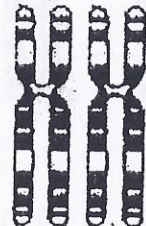
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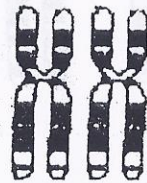
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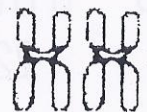
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