HUMAN CHROMOSOME SPREADS

KARYOTYPES AND MEDICAL GENETICS

Today, it is relatively easy to culture cells and obtain chromosome preparations. Usually, blood or amniotic fluid is obtained, and the cells are cultured for three days. During culturing, the drug **colchicine** is used to stop mitosis during **metaphase**. Thus, a large number of cells are arrested during this stage, when the chromosomes are the easiest to observe due to their extreme compactness. Each chromosome will have replicated (made a copy of) itself, and appears as two **chromatids** attached at the **centromere**. Cultured cells are then placed on a microscope slide, stained, and examined. One or more cells with all its chromosomes spread out and clearly visible is photographed. Once developed and printed, these photomicrographs of **chromosome (or metaphasic) spreads** are cut into individual chromosomes. The **karyotype** is built by taping or pasting these cutouts onto a karyotype form. Most modern cytogenetics laboratories now do all this work digitally on a computer.

Certain genetic disorders, such as Down Syndrome, Klinefelter Syndrome, Turner Syndrome, Cri-du-chat Syndrome, and numerous diseases can be diagnosed from the karyotype.

Your students will have the opportunity to assemble and interpret a number of karyotypes using **Human Chromosome Spreads.**

SUGGESTED TEACHER INSTRUCTIONS

1. Tell your students how metaphasic mitotic cells are prepared and photographed as described above.

2. Give each student a chromosome spread, a blank karyotype form, scissors, and tape or glue. We recommend **Human Chromosome Spread 1** (normal male) **or Spread 2** (normal female) to demonstrate the general principle of karyotyping. Students can then move on to play "genetic detective" with any of the abnormal spreads. You could give out as many different spreads as available so that the students are not analyzing identical spreads.

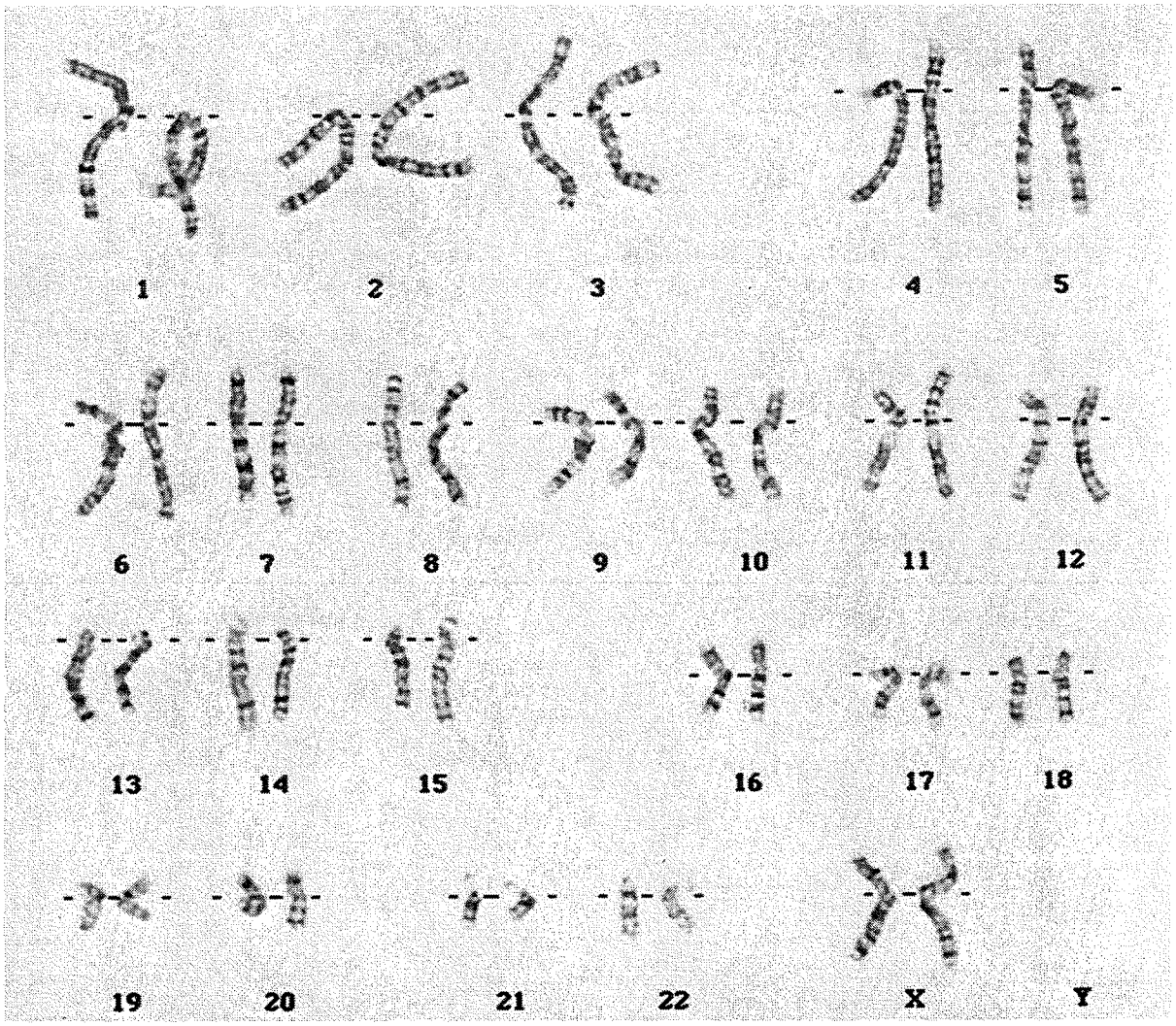
3. As a class, discuss the findings after the spreads are karyotyped. Your discussion should include what happens when the chromosomes are not "in balance" (too few, too many, translocated or deleted). Alternatively, have students conduct research on each disorder or distribute copies of **Human Karyotypes**, which give detailed information on each normal and abnormal karyotype.

IDENTIFICATION KEY

Spread 1: Normal male
Spread 2: Normal female
Spread 3: Down Syndrome male
Spread 4: Down Syndrome female
Spread 5: Klinefelter Syndrome XXY
Spread 6: Turner Syndrome X

Catalogue Number: 33W1057

Human Karyotype for
NORMAL FEMALE 46,XX



Normal females have 46 chromosomes, consisting of 22 pairs autosomes and two X chromosomes. In comparison to males, therefore. females have an extra copy of the X chromosome. In somatic tissue, one of the X chromosomes in each somatic cell is randomly selected to become condensed (forming the Barr body). Almost all of the genes in the condensed X chromosome are inactivated (a process also called "Lyonization"). The genes which escape inactivation appear to have diverse functions. Two copies of the X chromosome are required for normal ovarian function; deletion of all or part of an X chromosome may result in premature ovarian failure or ovarian dysgenesis.

Normal females produce haploid oocytes of the Karyotype 23,X. Oocytes frequently and randomly contain numerical and structural abnormalities of the chromosomes; most such abnormalities lead to miscarriage after conception.

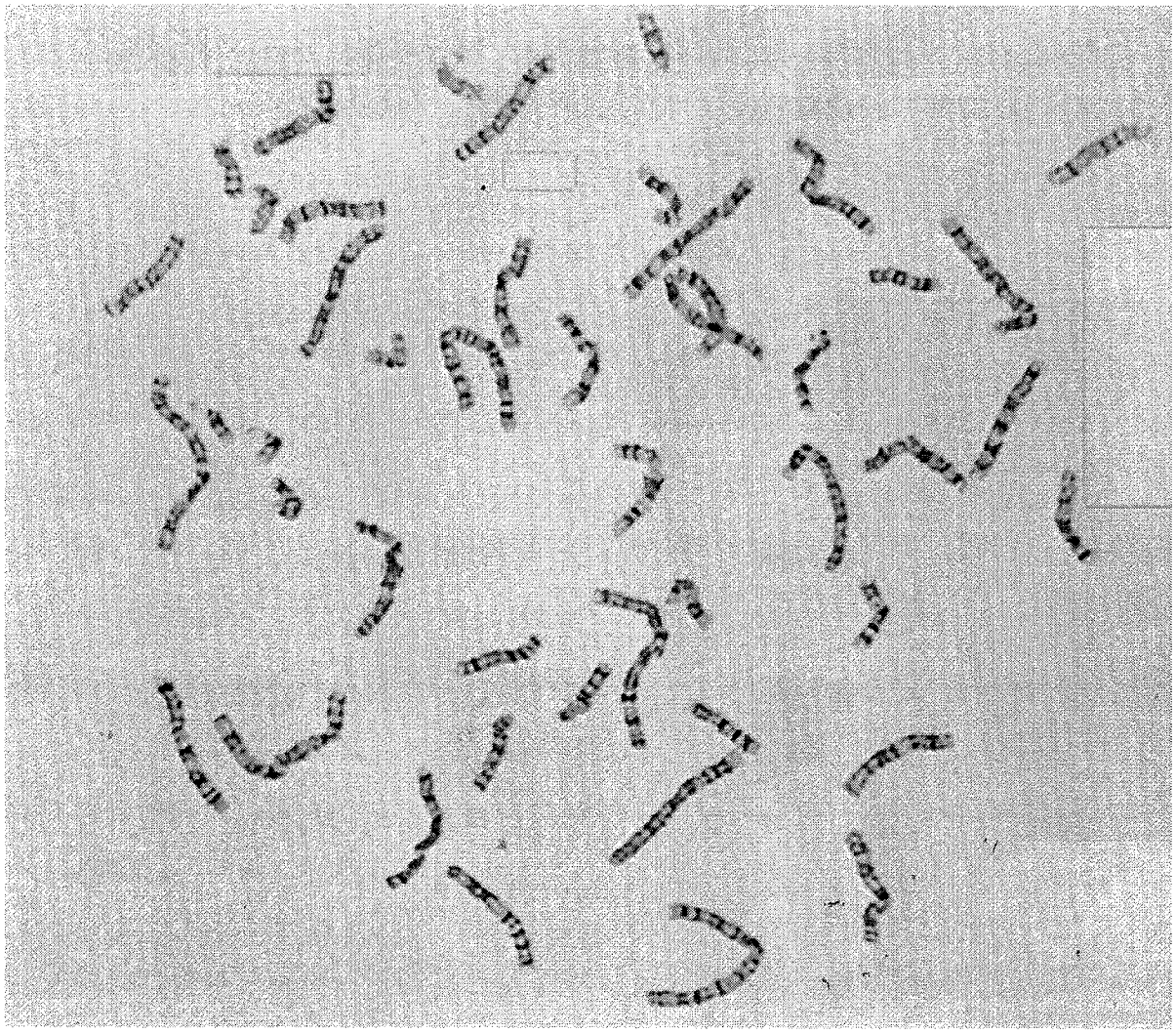
WARD'S

Catalogue Number: 33W1047

HUMAN CHROMOSOME SPREAD

The photomicrograph below is an enlarged picture of stained chromosomes in a single human cell treated with colchicine to arrest mitosis during metaphase. Cellular debris and organelles, such as the nucleolus, often appear in these photographs.

Spread 2



STUDENT INSTRUCTIONS FOR KARYOTYPING THIS SPREAD

1. Read the instructions carefully before beginning.
2. Using scissors, carefully cut out each chromosome.
3. Using a blank karyotype form, place each cutout chromosome onto the appropriate space on the blank form. Identifying the exact space for each chromosome is difficult at first. Note that the chromosomes are always arranged on the form in declining order of length with the exception of the X and Y chromosomes. The position of the centromere and the darkly stained bands also help identify each chromosome. There are two matching chromosomes at each numbered position in a normal individual. Chromosomes are frequently bent; this is not unusual or abnormal.
4. Once all the chromosomes have been classified on the karyotype form, tape or glue them in place.
5. At the bottom of the form, identify the number of chromosome and the sex if possible (XX = female, XY =male).
6. Is there any abnormality present? e.g. too many chromosomes, too few, parts missing, extra pieces stuck on one chromosome.
7. OPTIONAL: If a disorder is present, find out its name, symptoms, treatment if any, cause of the abnormalities, and whether or not this individual's offspring could inherit this disorder.