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.

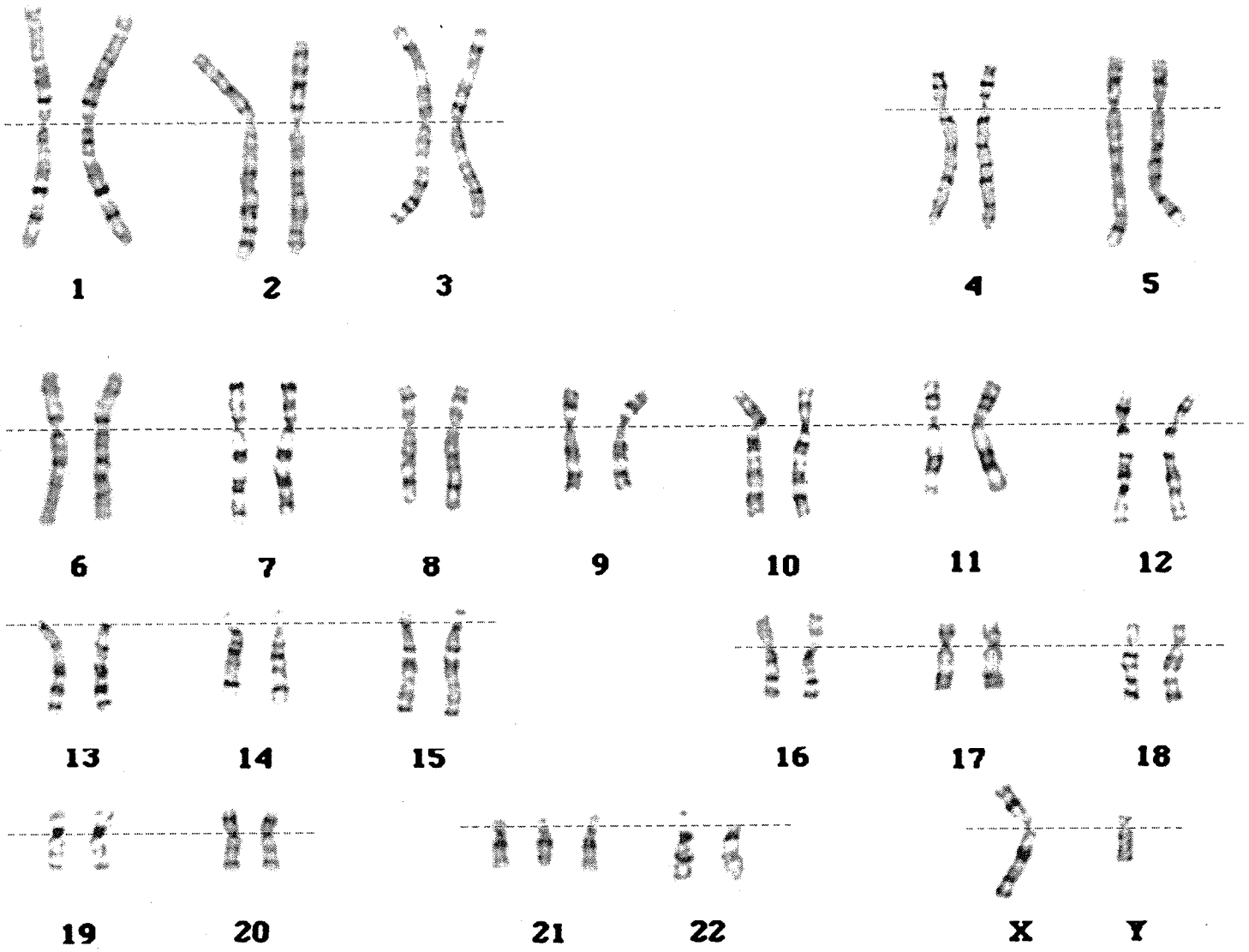
IDENTIFICATION KEY

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

WARD'S

Catalogue Number: 33W1058

Human Karyotype for
Down Syndrome, 47,XY,+21



More than 95% of all cases of Down syndrome are caused by the presence of an extra chromosome 21. Although the error may occasionally occur during spermatogenesis, trisomy 21 usually arises as a random error of oogenesis that is more likely to occur as the mother's age increases. The maternal age effect is substantial; at age 25, the risk for Down syndrome is approximately 1/1,200, while at age 35 it is approximately 1/260, and at age 45 it is approximately 1/25. The correlation between maternal age and the risk for Down syndrome has been the major basis for the development of cytogenetic tests in prenatal diagnosis (e.g. amniocentesis, chorionic villus sampling, and percutaneous umbilical blood sampling). These diagnostic procedures are now supplemented by tests such as maternal serum screening and ultrasonography, so that a women's choice to have cytogenetic testing may depend on a risk estimate based upon a combination of screening tests including age, serum biochemistry, and ultrasound evaluation.

Down syndrome is associated with numerous anomalies, of which the most common and serious are heart, kidney and intestinal malformations, susceptibility to respiratory infections, markedly increased risk of leukemia (especially in early childhood), poor muscle tone, and a moderate to severe decrease in intelligence with serious learning and behavioral anomalies. The face has a characteristic appearance, including a flattened nasal bridge, fold of skin at the corners of the eyes, and a large, protruding tongue. The potential for functional interaction in society depends in part upon supportive physical and educational therapy, but life expectancy is significantly shortened, and most people with Down syndrome cannot function independently in society. Males with Down syndrome are usually infertile.

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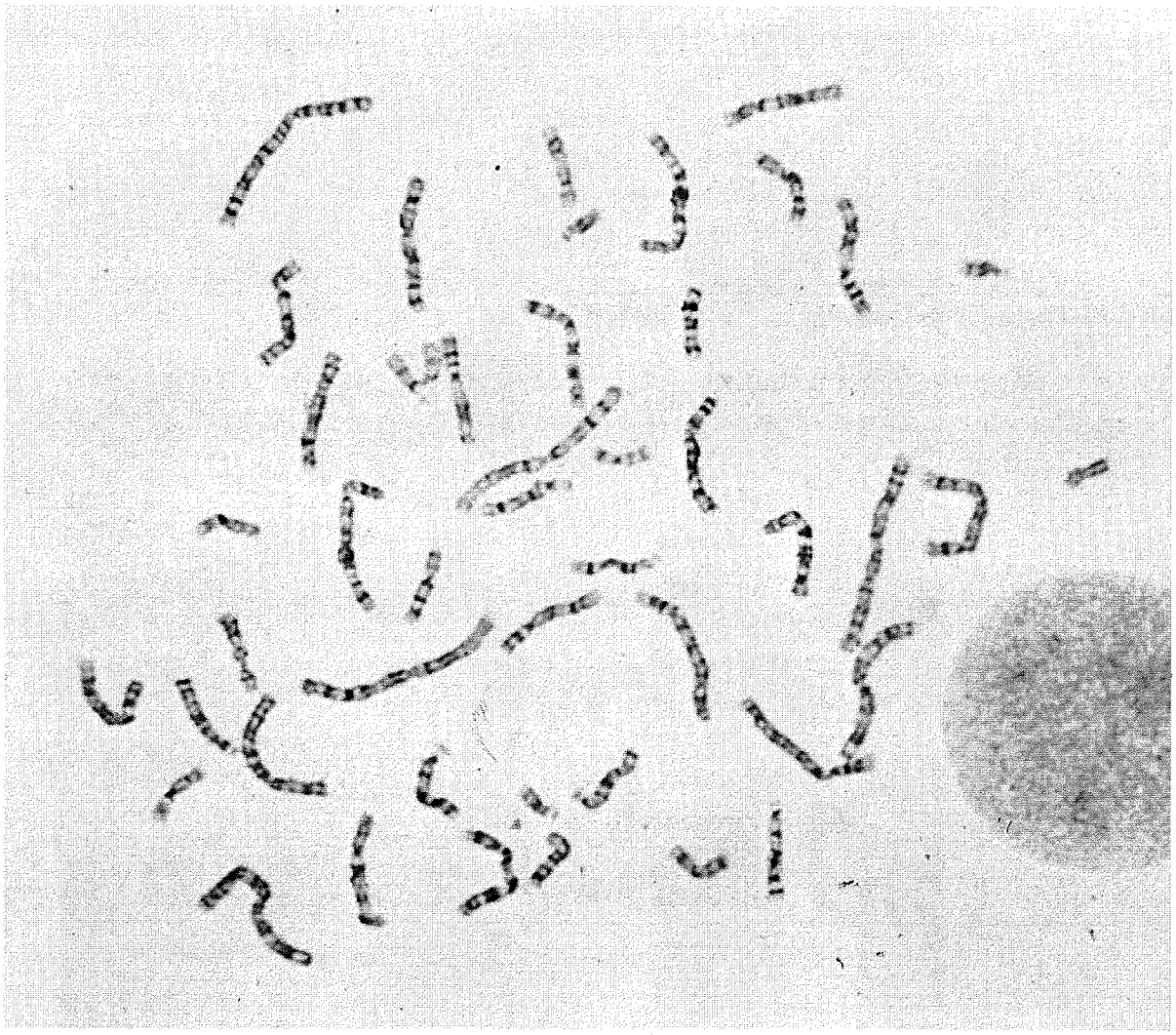
HUMAN CHROMOSOME SPREAD

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 3



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.