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

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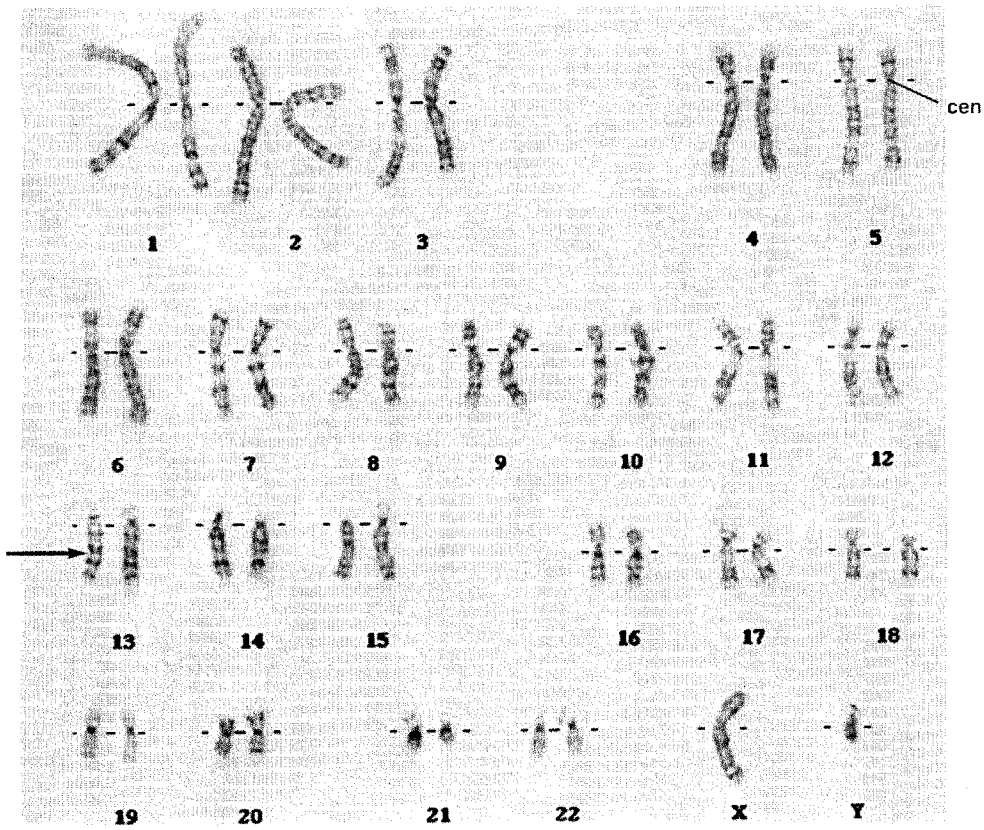
HUMAN KARYOTYPE FORM



Human karyotypes are built by arranging the chromosomes in declining order of size. Seven distinct groups (A to G), plus the sex chromosomes (X and Y) are the result. Besides length, the position of the centromere (**cen**), and the pattern of the bands (➔) help identify each chromosome. Use the example of the normal male karyotype to help you identify individual chromosomes in order to place them in the spaces below.

_____ _____ _____ _____ _____

1 2 3 4 5

Group A: Very long chromosomes: centromere in center

Group B: Long chromosomes; centromeres away from centre

_____ _____ _____ _____ _____ _____ _____

6 7 8 9 10 11 12

Group C: Medium length chromosomes; centromeres away from centre

_____ _____ _____ _____ _____ _____

13 14 15 16 17 18

Group D: Medium length chromosomes; Centromeres at or very near end

Group E: Chromosomes somewhat short; Centromeres away from centre

_____ _____ _____ _____ _____ _____

19 20 21 22 X Y

Group F: Short chromosomes; centromeres in centre

Group G: Very short chromosomes; centromeres at or near end

Sex Chromosomes

Number of Chromosomes: _____ **Sex of Subject:** _____ **Type of Disorder (if any)** _____

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