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The karyotype in Figure 1 is normal. The karyotypes in Figures 3 and 4 show evidence of an extra chromosome, while the karyotype in Figure 5 shows that a chromosome is missing. 2.

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A karyotype is an organized profile of a person's chromosomes. Two chromosomes specify sex, XX for female and XY for male. The rest are arranged in pairs, numbered 1 through 22, from largest to smallest.

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Chapter 14 The Human Genome Making Karyotypes Introduction Several human genetic disorders are caused by extra, missing, or damaged chromosomes. In order to study these disorders, cells from a person are grown with a chemical that stops cell division at the metaphase stage. During metaphase, a chromosome exists as two chromatids attached at the centromere.

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Karyotypes can also reveal the gender of a fetus or test for certain defects through examination of cells from uterine fluid – a procedure called amniocentesis – or through sampling of placental membranes. Over 400,000 karyotype analyses are performed each year in the U.S. and Canada.

### **Human Karyotyping Lab - University of Notre Dame**

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You will be completing a karyotype for Patient A, B & C Patient A ( Click on the link to "Complete Patient A's Karyotype" ) \*Match the chromosome to its homolog.

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Karyotypes are used by cytogenetic technologists, medical geneticists, or genetic counselors to determine whether a diagnosed birth defect is due to a chromosomal abnormality or to predict whether a fetus may be at risk for a genetic disorder.

## **Lesson 2: Analyzing Chromosomes**

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