2.3 Karyotyping

How do we use Karyotypes to help us learn about our traits?

Karyotyping is one of many techniques that allow us to look for several thousand possible genetic diseases in humans. During mitosis, chromosomes condense and are visible with a light microscope. A karyotype analysis usually involves freezing cells in mitosis and staining the condensed chromosomes with dye. The dye stains regions of chromosomes that are rich in the base pairs Adenine (A) and Thymine (T) producing a dark band.



The analysis involves comparing chromosomes for their shape, length, and the location and sizes of dyed bands. You will electronically complete the karyotype for three individuals and look for abnormalities that could explain the phenotype.

Karyotype notation:

Sex Chromosomes

Total # of Chromosomes Location of Extra or Missing Chromosome

Directions:

- 1. Go to the website <u>http://www.biology.arizona.edu/human_bio/activities/karyotyping/karyotyping.html</u>
- 2. Click on Patient Histories at the bottom.
- 3. Complete each patient's Karyotype (A, B, & C)
- 4. Complete the table below

Case	Karyotype Notation	Diagnosis	Chromosomal Abnormality
Α			
В			
С			

In humans, each cell normally contains 23 pairs of chromosomes, for a total of 46. One set of 23 chromosomes comes from you mother's egg, and one set of 23 chromosomes comes from your father's sperm. Twenty-two of these pairs, called autosomes, look the same in both males and females. The 23rd pair, the sex chromosomes, differ between males and females. Females have two copies of the X chromosome, while males have one X and one Y chromosome. The 22 autosomes are numbered by size. The other two chromosomes, X and Y, are the sex chromosomes. The pictures below of human chromosomes lined up in pairs are called karyotypes.

Person A						Person B							
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1. Based on the karyotypes above, which sex is each person? Explain how you know.

Person A is a ______ because

Person B is a _____ because

2. What is a chromosome? Explain how and when they form in cells.

3. Explain why there are two of each chromosome in the karyotypes above.

4. Each chromosome contains hundreds of genes that code for proteins. How is the information for making proteins stored in the DNA molecule? What happens if we have extra chromosomes, or if we are missing chromosomes?

5. Why might we get extra chromosomes?