

## Karyotypes: Diagnosing Chromosome Disorders

### HASPI Medical Biology Lab 17

#### Background/Introduction

#### DNA, Genes, and Chromosomes

All cells contain genetic information in the form of **deoxyribonucleic acid (DNA)** molecules. DNA contains the genetic instructions that form a species' characteristics. DNA is made up of four nucleotides: adenine, thymine, guanine, and cytosine. The arrangement, or order, of these nucleotides creates the directions for a specific protein. **Genes** are regions in the DNA that contain the instructions that code for the formation of proteins.

The Hierarchical Structure of DNA through to the Chromosome



Humans have more than 20,000 genes. All cells in an organism have the same genetic content, but the genes used (expressed) by the cell may be regulated in different ways. For example, the gene to create the actin and myosin proteins that are capable of contracting are expressed in muscle cells, but are dormant (not expressed) in skin cells. Not all DNA codes for a protein. Some segments of DNA are involved in regulatory or structural functions, and some have no as-yet known function.

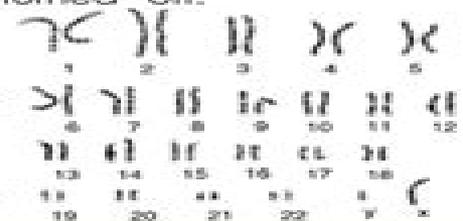
In each cell, DNA, and therefore genes, are wound into tight structures called **chromosomes**. Chromosomes are primarily formed during mitosis, or cell division, to prevent damage or loss of delicate DNA strands. Each chromosome consists of a single, very long DNA molecule. Humans have 23 pairs of chromosomes, 22 pairs of **autosomes**, and 1 pair of **sex chromosomes**. As their name implies, sex chromosomes contain the genes that primarily impact male and female characteristics.

#### Gene Expression and Regulation

How does a cell know when to express a gene? Some genes are always "on," and are responsible for cell processes such as metabolism, repair, and protein synthesis. Other genes are regulated and have the ability to be turned "on" and "off." For example, the gene that codes for insulin is only turned "on," or expressed, by pancreatic cells when it receives a signal from the brain. The brain only sends the signal when it senses a high level of glucose in the blood. As soon as the glucose level in the blood drops, the brain sends a signal to the pancreatic cells to stop using the gene for insulin, and the gene is turned "off."

#### Karyotypes

A genetic mutation that occurs when chromosomes do not separate correctly during cell division can be observed by looking at human DNA when it is in chromosome form. Observation can determine if there are any extra chromosomes, or missing or damaged chromosomes. This is done by removing the chromosomes from a cell, spreading them out, and making them visible since they are microscopic. A picture is then taken and a



# Chapter 1 human Chromosomes Answer Key

**Brendan G. Carr**



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