# WHAT IS GENETICS?

**Genetics** is the study of heredity. **Heredity** is how information is passed, or inherited, from parent to offspring. When an organism reproduces, it passes information to its offspring. This information codes for characteristics of the organism, including what the organism looks like and how it functions. All living things (including humans) contain this information. The information is stored in a molecule called DNA. **DNA** is often called the "blueprint for life" because it contains instructions for **traits**, or characteristics of organisms. DNA is found in the nucleus of a cell in humans and other eukaryotic organism. DNA is found in the cytoplasm in prokaryotic organisms, like bacteria.

**Chromosomes**

In humans and other eukaryotic organisms, DNA is separated into pieces. We call each one of these pieces a chromosome. A **chromosome** is a long, linear piece of DNA. The number of chromosomes found in a body cell of an organism is called the species chromosome number. A human body cell has 46 chromosomes. Other eukaryotic organisms have a different number of chromosomes. For example, a dog body cell has 48 chromosomes and a fruit fly body cell has 8 chromosomes. The species chromosome number is the same from generation to generation for a species. So, every human, from generation to generation, has 46 chromosomes in each of its body cells.

Every eukaryotic organism has pairs of chromosomes. We call these pairs of chromosomes **homologous chromosomes**. For example, a human has 46 chromosomes or 23 pairs of homologous chromosomes. Eukaryotic organisms have pairs of chromosomes because they reproduce sexually. **Sexual reproduction** means that an organism is produced from two parents. Each parent contributes one set of chromosomes to its offspring. For example, a human male passes 23 chromosomes and a human female passes 23 chromosomes to the offspring.

**Genes**

Each chromosome contains genes. A **gene** is a segment of a chromosome that codes for a specific trait. Genes are found at specific locations along a chromosome. Genes code for traits such as hair color, eye color and skin color. A chromosome can contain several hundred genes! Because eukaryotic organisms have pairs of chromosomes, they must have pairs of genes. We call a pair of genes, alleles. An **allele** is a form of a gene that controls a trait. So, every eukaryotic organism has two forms of a gene, alleles, that control a trait. Alleles are found at the same position along homologous chromosomes. What is important for you to understand now is that there are two copies of every gene in a eukaryotic organism.

# HUMAN HEREDITY

**Meiosis** is a type of cell division that results in four daughter cells each with HALF the number of chromosomes of the parent cell. The parent cell is **diploid** containing two sets of chromosomes. The resulting daughter cells are **haploid** containing only one set of chromosomes.

**Egg and Sperm**

There is one cell in females and one cell in males that have half the amount of genetic information as all of the body cells. These cells are called **gametes** or reproductive sex cells. Females produce a gamete called an egg. Males produce a gamete called sperm. In females, the egg is produced in the ovary. The ovary is an organ in the reproductive system. Females have two ovaries in their reproductive system. In males, sperm is produced in the testes. The testes are a pair of organs found in the male reproductive system. Eggs and sperm are produced by meiosis. The diploid body cells in humans have 46 chromosomes; therefore, the haploid gametes produced by meiosis have 23 chromosomes each.

**Meiosis**

In meiosis during gamete formation, the alleles for each gene segregate from each other, so that each gamete carries only one allele for each gene. This is called segregation. **Segregation** occurs due to the separation of homologous chromosomes in anaphase I of meiosis followed by the separation of sister chromatids in anaphase II of meiosis.

In addition, alleles for different genes usually segregate independently during the formation of gametes. This is called independent assortment. **Independent assortment** occurs because how the tetrads of different chromosomes line up in metaphase I of meiosis is random.

**Human Reproduction**

To produce a human offspring, the sperm and egg must unite. The sperm and egg fuse during a process called **fertilization**. When the sperm and egg fuse, the resulting cell is called a zygote. A zygote contains 46 chromosomes – 23 chromosomes from the egg and 23 chromosomes from the sperm.

The zygote will divide and divide until a cluster of cells forms. This type of cell division is called **mitosis**. All of these cells are the same as the original zygote. The cluster of cells is made of **undifferentiated** cells. In other words, all of these cells are un-different or NOT different. This cluster of cells eventually forms an **embryo**. During fetal development, the embryo develops into a baby with all the tissues, organs and organ systems of a human. This happens because of a process called **differentiation**. During differentiation, an embryo develops into a cluster of differentiated cells. In other words, the cells become different or specialized.

These cells develop into all of the different cells in the human body. Some of the cells will become bone cells. Others become muscle cells or skin cells. Different human body cells have different roles or jobs but they all contain the same 46 chromosomes. This is because each type of cell only uses part of the genetic information encoded in the chromosomes.

# MENDELIAN GENETICS

Much of today's knowledge of genetics and inheritance was a result of the research conducted by Gregor Mendel. Mendel was a scientist who lived during the 1800s. He was the first scientist to study heredity and is often called the "father of genetics." Surprisingly, all of his work was completed without knowledge of DNA or genes! It was not until the early 1900's that scientists observed chromosomes with a microscope.

Mendel studied pea plants and determined that traits are inherited in certain patterns. He studied pea plants because they had contrasting traits (tall and short, yellow and green seeds), are easy to grow and produced large numbers of offspring in a short amount of time.

**Mendel's Pea Plant Experiments**

Mendel grew pure-bred plants. Pure-bred plants produce offspring with the same characteristics as the parent plants, generation after generation. To determine how traits were passed from parent to offspring, he took purebred plants with contrasting traits and cross-pollinated them (took pollen from one plant and transferred it to the other plant).

In the first part of his experiment, Mendel took pollen from a tall purebred plant and transferred it to a short purebred plant. All of the offspring were hybrid tall plants. **Hybrids** are offspring of parents that have different traits. He called the offspring the $F_1$ generation.

In the second part of the experiment, Gregor Mendel cross-pollinated the $F_1$ plants. The offspring of the $F_1$ plants were called $F_2$ plants. Unlike the results of the $F_1$ generation, some plants were tall and others were short. How could this be?

Mendel determined that some traits are "stronger" than other traits. He called these traits dominant. **Dominant** traits always appear when present. The trait that is hidden by the dominant trait is called the **recessive** trait. As a result of modern genetic research, we now know there are two copies of every gene for every trait.

There are three possible combinations of genes because organisms have two copies of every gene for every trait. An organism can have: 2 dominant alleles; 2 recessive alleles; or 1 dominant allele and 1 recessive allele. If an organism has two of the same alleles, it is called **homozygous**. An organism with two dominant alleles for a trait is homozygous dominant for that trait. An organism with two recessive alleles for a trait is homozygous recessive for that trait. An organism with two different alleles for a trait is **heterozygous**. In this case, the dominant allele will be expressed and the recessive allele will be hidden.

Not all traits are controlled by one gene with two alleles. Some genes have multiple alleles and some traits are controlled by more than one gene. Furthermore, some alleles are not entirely dominant or recessive.

**Genotype vs. Phenotype**

Scientists study genes and traits differently. Sometime scientists are only concerned about the **phenotype**, the physical makeup or appearance of an organism. Other scientists study the **genotype**, the genetic makeup of an organism. The phenotype of an organism is determined by the genotype of the organism. In other words, the appearance or physical makeup of the organism is determined by the genetic makeup of the organism.