**An Overview of Genetics**

**Introduction**

If anyone has ever told you that you have your mother’s eyes, or that your athletic ability runs in your family – they were probably onto something. **Genetics** is a field of study that focuses on study of **heredity**, or how genetic information is passed from one generation to another.

Humans, along with every other living organism on Earth, share one common trait – they all contain genetic information. Every animal, plant, and bacterium is made up of **cells**.The human body is made of trillions and trillions of them. Inside each individual cell is a full set of genetic information, or a **genome**. The human genome is organized into **chromosomes**. Each chromosome contains hundreds, or even thousands, of **genes**. Genes are coding sequences that give a blueprint to each and every trait that the organism has. Gene sequences can be anywhere between a few hundred and several million units of **DNA**, the fundamental unit of genetic information. There are approximately 3 billion units, or **base pairs**, of DNA making up the human genome.

Understanding genetics allows us to understanding where we came from and what we’re made of. Our genetics determine everything about us, including how tall we are, what our voice sounds like, and how we think and interact with others. They also determine our health and can cause us to be more or less susceptible to illness and disease. The genetics field has been making major advancements in recent decades, and scientists hope to develop personalized medical treatments that can cure diseases and help us stay healthy.

**DNA and Genes**

DNA is made up of **base pairs**. There are four main nitrogenous bases that make up the human genome – adenine (A), thymine (T), guanine (G), and cytosine (C). Base pairs are formed by Adenine binding with Thymine and Guanine binding with Cytosine. Two opposite, complementary strands of DNA, held together by a sugar-phosphate backbone, are engineered to specifically bind to one-another via base pairs. These pairs connect the strands, forming a double helix structure.

If all of the DNA in one single human cell were to be unraveled and laid out in a straight line, it would measure approximately six feet long. In order to fit all of this into one tiny compartment, the DNA strands must be condensed extremely tightly. To do this, group of specialized proteins called **histones** wrap themselves in and around DNA. They condense into structures called chromatin, which is how DNA exists most of the time. During cell division, in order to distribute DNA to its correct location, this chromatin further condenses into structures called **chromosomes**. Human DNA is split into 46 strands that condense into 46 chromosomes, or 23 chromosome pairs.

Not all of our DNA is part of a gene. In fact, only about 1.5% of the genome is part of a coding sequence. The remaining 98.5% was once believed to serve no purpose, but many scientists now believe that at least some of it is involved in regulating gene expression and activation.

**Genotype Versus Phenotype**

A set of human DNA contains 23 chromosome pairs, and 46 chromosomes total. The two chromosomes in a pair contain the exact same genes in the exact same locations. However, the specific gene sequences may not be identical, and they may code for different products. **Alleles** are alternate variations of genes that you can inherit from each parent. Besides the X and Y chromosomes, which are sex-specific, chromosome pairs have the same genes, but may have different alleles.

Our **genotype** refers to the collection of all the alleles we carry for a trait, whether we express them or not. **Phenotype**, on the other hand, only includes observable traits. Consider a puppy. If its mother had brown fur and its father had black fur, the puppy’s genotype would contain alleles for both brown and black fur. However, if the puppy had brown fur, its phenotype would only include the allele for brown fur.

**Genetic Inheritance**

**Heredity** is the inheritance, or passing, of genes from parents to offspring. Gregor Mendel, sometimes called the “Father of Genetics”, is responsible for discovering that each trait has a gene, and each gene has two alleles. He determined this by observing pea plant phenotypes in the 1860s. His work and fundamental laws of genetic inheritance still hold up today.

One of Mendel’s most significant works was his discovery of dominant and recessive inheritance. A **dominant** allele produces a dominant phenotype, meaning that the presence of the allele on only one gene copy is enough for the trait to be phenotypically expressed. A **recessive** allele, on the other hand, must be present in both gene copies in order for its respective trait to be expressed.

Organisms who have only one recessive copy of an unexpressed gene in their genotype are called **carriers**. Carriers can pass their recessive allele to their offspring, but the trait will not be expressed unless both parent organisms pass on the recessive allele and the offspring receives two copies of it. If both parents are carriers, the offspring will have a 75% chance of being a carrier and a 25% chance of expressing the trait.

**Genetic Mutations and Diseases**

Considering how many genes we have and how much DNA is in each one, it’s a wonder how we aren’t filled with **mutations**, or changes in the DNA sequence that result in different sequences than they were supposed to. As our DNA is being constructed in our cells, it is checked and re-checked to make sure there are no mistakes. However, some mutations do get past the proofreading molecules and change a part of the cell’s DNA. Mutations can be hereditary or somatic, meaning they can be passed down from a parent to an offspring or can develop later in life.

Mutations can affect just one single gene, like in Cystic Fibrosis, or they can affect entire chromosomes, like in Down Syndrome. Some genes’ purpose is to send signals to other cells to divide. If a mutation causes these genes to constantly be activated, cells can start to divide rapidly and turn into tumors and cancers. Cancer can also be promoted by a mutation in a cell’s proofreading machinery.

Many genetic diseases have recessive patterns of inheritance, which is what makes them so rare. However, it is not uncommon for an individual to be a carrier for multiple genetic diseases or mutations. Carriers usually have no idea unless that they have an allele for a disease unless they are screened, because the dominant gene copy is expressed instead.

If two parents happen to be carriers for the same recessive genetic disease allele, their offspring will have a 3 in 4 chance of being a carrier and a 1 in 4 chance of actually expressing the disease. Couples interested in having children can obtain a carrier screening to determine if they carry any of the same recessive alleles.

**The Human Genome**

The human **genome** is the entirety of all the genetic information within a complete set of DNA. **Genomics** is the field of study that focuses on understanding the human genome and its contents. Genomics have been a hot-topic in scientific research in recent decades and there have been substantial scientific advancements that have taught us more about what makes us, us. Arguably the most substantial advancement was the completion of the Human Genome Project, an international project that set out to make a map of the entire human genome and identify the locations of genes. Prior to this, scientists had a general idea of the structure of chromosomes and the placement of genes, but they wanted a deeper and more comprehensive understanding of the “big picture”. The 15-year study took DNA samples from thousands of participants and analyzed them using DNA sequencing techniques. Eventually, they were able to come up with a full reference set of DNA, or a map of the human genome. Using the reference genome, individuals can now have their own genome mapped and compared to the reference set to identify genetic abnormalities or susceptibilities they may carry.

There is still much more to learn about the human genome, but completing this project was a huge accomplishment in the genetic field and has opened the door to many possibilities. In the future, the results of the project could help lead to medical and pharmacological procedures and treatments that were once thought to be impossible, including treating and curing genetic diseases and developing genetic engineering methods.

**Conclusion**

In this chapter, we covered a very broad scope of material, and each component of genetics is found to be more and more complex the more it is studied. However, a basic understanding of genetics and heredity can help us understand why and how we are who we are. Genetic research is progressing further every day, and in the future, it could save many lives. Our bodies are astonishingly complex, and we, as humans, are just beginning to understand them.

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