Basic Genetics

Genetics is the study of biologically inherited traits determined by elements of heredity that are transmitted from parents to offspring in reproduction. These inherited elements are called **genes**. Recent advances in the field of **genomics** have led to development of methods that can determine the complete **deoxyribonucleic acid (DNA)** sequence of an organism. Genomics is the latest advance in the study of the chemical nature of genes and the ways that genes function to affect certain traits.

The work of Gregory Mendel, a monk and part-time biologist, with garden peas is regarded as the beginning of what would become the science of genetics. Mendel is credited with showing the existence of genes as well as illuminating the rules governing their transmission from generation to generation. The study of genetics through the analysis of offspring from mating is sometimes referred to as classical genetics.

Heredity is the cause of similarities between individuals. This is the reason that brothers and sisters with the same parents resemble each other and with their parents. **Variation** is the cause of differences between individuals. This is the reason that brothers and sisters who do resemble each other are still unique individuals. **So** The heredity and variations play an important role in the formation of new species (speciation).

The billions of nucleotides in the nucleus of a cell are organized linearly along the DNA double helix in functional units called genes. Each of the 25,000 to 30,000 human genes is accompanied by various regulatory elements that control when that gene is active in producing **messenger ribonucleic acid (mRNA)** by the process of **transcription**. In most situations, mRNA is transported from the nucleus to the cytoplasm, where its genetic information is used in the manufacture of proteins (a process called **translation)**; these proteins perform the functions that ultimately determine phenotype.

For example, proteins serve as enzymes that facilitate metabolism and cell synthesis; as DNA binding elements that regulate transcription of other genes; as structural elements of cells and the extracellular matrix;

and as receptor molecules for intracellular and intercellular communication. DNA also encodes many small RNA molecules that serve functions that are not yet fully understood, including regulating gene transcription and interfering with the translational capacity of some mRNAs.

Chromosomes are the means by which the genes are transmitted from generation to generation. Each chromosome is a complex of protein and nucleic acid in which an unbroken double helix of DNA is tightly wound. Genes are found along the length of chromosomes. A variety of highly complicated and integrated processes occur within the chromosome, including DNA replication, recombination, and transcription.

In the nucleus of each of their somatic cells, humans normally have 46 chromosomes, which are arranged in 23 pairs. One of these pairs, consisting of the **sex chromosomes** X and Y, determines the sex of the individual; females have the pair XX, and males have the pair XY. The remaining 22 pairs of chromosomes are called **autosomes.** In addition to these nuclear chromosomes, each mitochondrion (an organelle found in varying numbers in the cytoplasm of all cells) contains multiple copies of a small chromosome. The **mitochondrial chromosome** encodes a few of the proteins for oxidative metabolism and all of the **transfer ribonucleic acids (tRNA)** used in translation of proteins within this organelle. Mitochondrial chromosomes are inherited almost entirely from the cytoplasm of the fertilized ovum and, therefore, are maternal in origin.

The exact location of a gene on a chromosome is known as its **locus**, and the array of loci constitutes the human gene map. Currently, researchers have identified the chromosomal sites of more than 11,000 genes (i.e., those for which normal or abnormal function has been identified).

Homologous copies of a gene are termed **alleles**. In comparing alleles, it must be specified at which level of analysis the comparison is being made. For example, if alleles are truly identical, their coding sequences and the number of copies do not vary, so the individual is **homozygous** at that specific locus.

However, if the DNA is analyzed using either restriction enzyme examination or nucleotide sequencing, then, despite having the same functional identity, the alleles would be viewed as different, and the individual would be **heterozygous** for that locus. Heterozygosis based on differences in the protein products of alleles has been detectable for decades and represents the first hard evidence proving the high degree of human biologic variability. In the past decade, analysis of DNA sequences has shown genetic variability to be much more common, with differences in nucleotide sequence between individuals occurring about once every 1,200 nucleotides.

The study of the mode of gene transmission from generation to generation is broadly called **transmission genetics**; the study of structure and function of the gene forms the **molecular biology**.