The Physical Basis of Heredity

GENETICS

Genetics is that branch of biology concerned with heredity and variation. The hereditary units that are transmitted from one generation to the next (inherited) are called genes. The genes reside in a long molecule called deoxyribo nucleic acid (DNA). The DNA, in conjunction with a protein matrix, forms **nucleoprotein** and becomes organized into structures with distinctive staining properties called chromosomes found in the nucleus of the cell. The behavior of genes is thus paralleled in many ways by the behavior of the chromosomes of which they are a part. A gene contains coded information for the production of proteins. DNA is normally a stable molecule with the capacity for self-replication.

On rare occasions a change may occur spontaneously in some part of DNA. This change, called a **mutation,** alters the coded instructions and may result in a defective protein or in the cessation of protein synthesis. The net result of a mutation is often seen as a change in the physical appearance of the individual or a change in some other measurable attribute of the organism called a **character or trait.** Through the process of mutation a gene may be changed into two or more alternative forms called allele morphs or alleles.

Example: Healthy people have a gene that specifies the normal protein structure of the red blood cell pigment called hemoglobin. Some anemic individuals have an altered form of this gene, i.e., an allele, which makes a defective hemoglobin protein unable to carry the normal amount of oxygen to the body cells.

Each gene occupies a specific position on a chromosome, called the gene **locus (loci,** plural). All allelic forms of a gene therefore are found at corresponding positions on genetically similar **(homologous)** chromosomes. The word "locus " is sometimes used interchangeably for "gene. " When the science of genetics was in its infancy the gene was thought to behave as a unit particle These particles were believed to be arranged on the chromosome like beads on a string. All the genes on a chromosome are said to be linked to one another and belong to the same **linkage group.** Wherever the chromosome goes it carries **all** of the genes in its linkage group with it. As we shall see later, linked genes are not

transmitted independently of one another, but genes in different linkage groups (on different chromosomes) are transmitted independently of one another.

What is a gene?

A gene is the basic physical and functional unit of heredity. Genes are made up of DNA. Some genes act as instructions to make molecules called proteins. However, many genes do not code for proteins on page 18. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. The Human Genome Project estimated that humans have between 25,000 and 30,000 genes. Every person has two copies of each gene, one inherited from each parent. Most genes are the same in all people, but a small number of genes (less than 1 percent of the total) are slightly different between people.

Alleles are forms of the same gene with small differences in their sequence of DNA bases. These small differences contribute to each person's unique physical features. Scientists keep track of genes by giving them unique names. Because gene names can be long, genes are also assigned symbols, which are short combinations of letters (and sometimes numbers) that represent an abbreviated version of the gene name. For example, a gene on chromosome 7 that has been associated with cystic fibrosis is called the cystic fibrosis trans membrane conductance regulator; its symbol is CFTR. Genes are made up of DNA and Each chromosome contains many genes.

How Genes Determine Traits

The connection between genes and traits is definitely not simple. Most visible traits of organisms are the net result of many genes acting together in combination with environmental factors. Therefore, the relationship between genes and traits is often complex for one or more of the following reasons:

1. One gene can affect more than one trait.
2. One trait can be affected by more than one gene.
3. Many traits are affected by environmental factors as well as by genes.

Genes consist of three types of nucleotide sequence:

1- coding regions, called exons, which specify a sequence of amino acids

2- non-coding regions, called introns, which do not specify amino acids

3- regulatory sequences, which play a role in determining when and where the protein is made (and how much is made)