Chapter 13 - Sources of Genetic Variation

The existence of HERITABLE (GENETIC) VARIATION is essential for evolution.

Unfortunately for Darwin, the predominant view of heredity during his time was that of BLENDING INHERITANCE - Heredity “stuff” of parents blend together to produce characteristics observed in the offspring.

The inheritance problem was solved by Mendel’s experiments with peas plants. His work showed that inheritance is PARTICULATE - Heredity factors from the parents (=genes) will remain unchanged in the hybrid.

In addition to genetic variation, for evolution to proceed there must be a fairly continuous origin of new genetic variation.

Q. How does genetic variation arise?

The ultimate source of genetic variation in populations is via MUTATION.

Types of Mutations

There are two general types of mutations: point (gene) mutations and chromosomal mutations.

Point mutations affect only one or a few nucleotides within a gene.

Chromosomal mutations change the number of chromosomes or the number or arrangement of genes in a chromosome (= change in chromosome structure).

Point (Gene) Mutation

A POINT MUTATION is a change in one nucleotide or a few nucleotides in a single gene. Point mutations within a gene can be divided into two general categories: base pair substitutions and base pair insertions or deletions.
**Base Pair Substitutions**

A base pair substitution is the replacement of one nucleotide, and its partner from the complimentary DNA strand, with another pair of nucleotides.

Some substitution mutations have no effect on the protein coded for.

There are at least four reasons for this:

1. Because of the redundancy of the genetic code

Substitutions are often referred to as **misense mutations**

2. A change in an amino acid may have no effect on a protein’s function.

There are regions in proteins that are sensitive to amino acid changes and some that are relatively insensitive.

3. Third, changes in introns (noncoding) will have no effect because these regions are not translated.

4. And fourth, a change in a gene may be masked by the presence of other, normal copies of the gene.

A point mutation (base pair substitution) can, however, have important consequences.

One example is a change that produces one of the chain-terminating, or stop codons (UAA, UAG, or UGA), such as a transition from **UAC** (serine) to **UAA** (stop).

Alterations that change an amino acid codon to stop a signal are called **nonsense mutations**

If this occurs, translation is halted before the entire protein is translated, potentially causing a major change in the structure and function of the protein.

**Insertions and Deletions**

Insertions and deletions are additions or losses of one or more nucleotide pairs in a gene.

They can cause disruption of the normal 3 nucleotide reading frame, hence the name **frameshift mutation**
Chromosomal Mutations
A change in the number of chromosomes or arrangement of genes in chromosomes

Changes in the structure of a chromosome (4)

a. **DELETIONS** - a chromosome segment is lost from a chromosome

b. **DUPLICATIONS** - A chromosome segment is present more than once in a set of chromosomes
Both of these generally result from **UNEQUAL CROSSING OVER** during synapsis in meiosis

c. **INVERSIONS** - result when two breaks occur in a chromosome and the broken segment is rotated 180°.
If the inverted segment includes the centromere, the inversion is called **PERICENTRIC INVERSIONS**, if not the inversion is **PARACENTRIC INVERSIONS**

d. **TRANSLOCATIONS** - The location of a chromosome segment is changed
The most common forms of translocations are **reciprocal** - result from the exchange of segments from two non-homologous chromosomes (two chromosomes that carry different genes)
But a chromosomal segment may also move to a new location within the same chromosome, or in a different chromosome without reciprocal exchange
These kinds of translocations are called **transpositions**

2. Changes in the number of chromosomes

a. **Centric fusion** - Two nonhomologous chromosomes fuse into one
This entails a loss of a centromere

b. **Centric fission** - One chromosome splits into two

c. **Aneuploidy** One or more chromosomes of a normal set are lacking or present in excess
e.g. trisomic - occurrence of chromosomes 3 times
It is due to **nondisjunction** - members of pairs of homologous chromosomes do not separate during meiosis I or sister chromatids fail to separate during anaphase of meiosis II.

**d. POLYPLOIDY** - the duplication of chromosome sets such that individuals have more than 2 of each chromosome.  
**DIPLOIDY** is the normal state (2 of each chromosome, 2N), but some animals are **TRIPLOID** (3N) and **TETRAPLOID** (4N).

There are 2 types of polyploidy

1. **Autopolyploidy**

   The multiplication of chromosomes sets within a species  
   For example, a failure of meiosis during gamete production can double chromosome number from the diploid count (2N) to a tetraploid number  
   Occurs due to **nondisjunction** - an accident during meiosis (e.g. during gamete formation) in which homologous chromosomes fail to separate  
   
   The tetraploids formed can mate with themselves (self-pollinate) or with other tetraploids  
   However, the tetraploid mutants cannot successfully interbreed with diploid individuals from the original population  
   The offspring would be sterile triploids (3N); sterile because unpaired chromosomes result in abnormal meiosis

2. **Allopolyploidy**

   A much more common type of polyploidy  
   It refers to the contribution of two different species to the polyploid hybrid  
   It begins with 2 different species interbreeding and combining their chromosomes  
   
   Interspecific hybrids are usually sterile because the haploid set of chromosomes from one species cannot pair during meiosis with the haploid set from the other species.
Maintaining Genetic Variation

Genetic variation is largely maintained during meiosis. During meiosis, paternal and maternal chromosomes are reshuffled and new chromosome combinations occur.

One way genetic recombination occurs is by independent assortment. Another meiotic mechanism that ensures variety in the gametes is called chromosomal crossing over.

Because gametes produced by meiosis are haploid, sexual reproduction is necessary to reconstitute the diploid genome. This recombination of gametes from genetically different individuals adds another layer to the number of possible combinations of alleles. With sexual reproduction, the total number of truly genetically unique individuals becomes staggering.

Conclusions

Without genetic variation, evolution cannot occur - no genetic variation = no evolution.

The origin of new genetic variation is predominantly through mutation. New alleles are produced primarily by point mutations. [point mutation --> new alleles]

An existing allele is hit by a mutation, and as a consequence, becomes a new allele. Of course most are deleterious; those that are not are usually recessive and thus masked. Mutations are then often free to accumulate on these extra copies, and eventually, these copies may produce proteins that are favored by selection.

Chromosomal mutations primarily move existing alleles around -- either from one location on a chromosome to another on the same chromosome or from one chromosome to a different chromosome. But chromosome mutations rarely, by themselves, produce new alleles. Gene duplications resulting from chromosome mutations can produce new genes; that is, new stretches of DNA. [gene duplication --> new genes]
Crossing over, independent assortment, and sex are the principle mechanisms that maintain genetic diversity within populations.

Point mutations

![Diagram of point mutations]

**Figure 16.23**
Categories and consequences of point mutations. Mutations are changes in DNA, but they are represented here as they are reflected in mRNA and its protein product.