

## Sources of Genetic Variation

As Darwin realized, the existence of **HERITABLE (GENETIC) VARIATION** is *essential* for evolution

Without heritable variation, any trait favored by natural selection will not be passed on to offspring.

If favorable traits are not passed on offspring, evolutionary change cannot occur.

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

Blending inheritance posed a major problem for Darwin

It could only result in a reduction of genetic variation.

Over time, a population would become more and more homogenous, and eventually, genetic variation would disappear.

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

Today we might say discrete alleles coding for variants of a trait are passed to offspring and maintain their integrity across generations

Thus, there is no loss of genetic variation with each successive generation.

So, with blending inheritance, variation is rapidly lost as extreme types mate together and their genes are blended out of existence in some general mean form

In Mendelian inheritance, variation is preserved because the extreme genetic types (even if disguised in the heterozygotes) are passed down from generation to generation

There was another issue concerning blending inheritance that was resolved with Mendelian heredity

Under blending heredity, a rare new advantageous gene is soon to be blended away

Under Mendelian heredity a rare new favorable gene can increase in frequency and eventually become established in the population

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.**

As a rule, the information encoded in the nucleotide sequence of DNA is faithfully reproduced during replication so that each replication results in two DNA molecules identical to each other and to the parental one

Occasionally, however, mistakes or mutations occur (1/billion nucleotides or 3/mitosis), so that daughter cells differ from the parental cells in the DNA sequence or in the amount of DNA

**Types of Mutations: A Review**

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

About 24% of base pair substitutions will code for the same amino acid

Substitutions are often referred to as **misense mutations** - the altered code still codes for an amino acid and therefore it still makes sense, although not necessarily the right sense  
In short, a change in a base pair may transform one codon into another codon that is translated into the same amino acid

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 tend to have much greater effects than do substitutions.

This is because they can cause disruption of the normal 3 nucleotide reading frame, hence the name **frameshift mutation**

But, if a deletion occurs in a multiple of 3, the structure of the resulting protein may be only slightly altered.

Deletions in multiples other than 3, however, can affect the entire structure of the protein.

### **Chromosomal Mutations**

Different cells of the same organism and different individuals of the same species have, as a rule, the same number of chromosomes

Homologous chromosomes, are as a rule, uniform in number and in the order of genes they carry

These rules have exceptions known as **CHROMOSOMAL MUTATIONS**.

A change in the number of chromosomes or arrangement of genes in chromosomes

So, there are 2 general categories of chromosomal mutations: changes in the number of chromosomes and changes in the structure of chromosomes

#### ***Changes in the structure of a chromosome (4)***

These chromosome mutations involve rearrangement of genetic material on the chromosomes

**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

As a result of unequal crossing over, one gamete receives a chromosome with a duplicated gene or genes while the other gamete receives a chromosome with a missing gene or genes.

While deletions are usually deleterious, duplications can be advantageous.

Duplication is also one of the primary ways that there can be increases in genome size. e.g. additional genes yield more complexity

**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**

One consequence of inversions is that they often either prevent crossing over or result in the products of crossing being eliminated during meiosis (in the handout notice that chromosomes pair by forming loops)

This allows alleles at a sequence of genes to be preserved together.

If two genes are near each other on a chromosome, and crossover products never occur, the alleles at these genes will always be transmitted together.

So if alleles at a series of genes work particularly well together, inversions can result in their being maintained together through many generations - **coadapted gene complexes**

**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**

Most translocations are deleterious, but there is clear evidence, however, that translocations have occurred and been perpetuated in a number of species

One consequence of a translocation is that it alters patterns of linkage. (quite possibly breaking up coadapted gene complexes)

Alleles that tended to be transmitted together because they were on the same chromosome can assort independently after a translocation.

## 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

A new centromere must be produced, otherwise the chromosome without the centromere would be lost when the cell divides

**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).

Polyploidy is common in plants (Nearly half of all angiosperm species are polyploid, as are a majority of ferns) and rare in animals (Occurs among hemaphroditic earthworms and flatworms)

Polyploidy is present in some parthenogenic animals (e.g., animals that can produce offspring without fertilization), including some species of beetles, goldfish, and salamanders

There are 2 types of polyploidy

### ***1. Autopolyploidy***

The multiplication of chromosome 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

This reshuffling during meiosis is called **genetic recombination**

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.