Fundamental bases for genetic diversity:
(after Falconer, 1960, Quantitative Genetics)

*Mutation*

*Migration*

Drift (*chance divergence in time/space*)

*Selection*
Genetic Mutations

Source of genetic variability and critical for evolution

- Classified as beneficial, harmful or neutral
  - Harmful mutations are lost if they reduce fitness of an individual
  - If fitness is improved by a mutation, then the frequency of that allele will increase from generation to generation
  - The mutation could be transparent: to resemble an allele already in the population
• A mutation could generate an entirely new allele.
  o Most of these mutations will be detrimental and lost.
  o If the environment changes, a new mutant allele may be favored and eventually become the dominant allele in that population.
  o If the mutation is beneficial to the species as a whole, migration must occur for it to spread to other populations of the species.

• Gene duplication - favors and facilitates mutational events.
  o The duplicated gene can undergo mutations to generate a new gene that has a similar, but a slightly modified function for the organism.
  o This type of evolution generates multigene families. (Examples: gluten proteins and other seed storage genes, photosynthetic genes in plants, disease resistance genes)
Chromosomal Mutations - *Large sections of chromosomes can be altered or shifted, leading to changes in the way the genes on them are expressed.*

- **Translocations** - involve the *interchange of large segments* of DNA between two different chromosomes.

- **Inversions** - occur when a region of DNA *flips its orientation* with respect to the rest of the chromosome. This can lead to the same problems as translocations.

- **Deletions** - Sometimes large regions of a chromosome are deleted. This can lead to a *loss of important genes*.

- **Nondisjunction** - Sometimes *chromosomes do not segregate properly* during cell division, especially if large segments are rearranged. One of the daughter cells will end up with more or less than its share of DNA.
Point mutations - *single base pair changes*.

- **A nonsense mutation** creates a stop codon (i.e. UAA, UGA, or UGG) where none previously existed. This shortens the resulting protein, possibly removing essential regions.

- **A missense mutation** changes the code of the mRNA. If an AGU is changed to an AGA, the protein will have an arginine where a serine was meant to go. This might **alter the shape or properties of the protein**.

- **A silent mutation** has no effect on protein sequence. If an AGU was changed to an AGC, the protein would still have the appropriate serine at that position.

- **Frame shift** from small deletions or insertions of bases not divisible by 3.
How / why do mutations occur??

Errors in cellular replication
   *May be induced by radiation, chemicals, or even environmental stress*

Transposons
   Mobile genetic elements that can move in and out of chromosomes, which can alter gene expression.

Transposons have been found in all organisms studied to date, and thus are an important force for generating diversity as a basis for evolution.

Barbara McClintock in the late 40s and early 50s developed the concept of movable genetic entities, which she initially termed "controlling elements". For her work McClintock received the Nobel Prize in 1983.
Mutations frequencies

Naturally occurring changes due to errors in DNA replication and repair:

Approx. 1 in $10^7$ to $10^8$

(1 in 100 million as based on bacterial studies)

*Mutation events that have a positive influence on fitness or survival are extremely rare!*