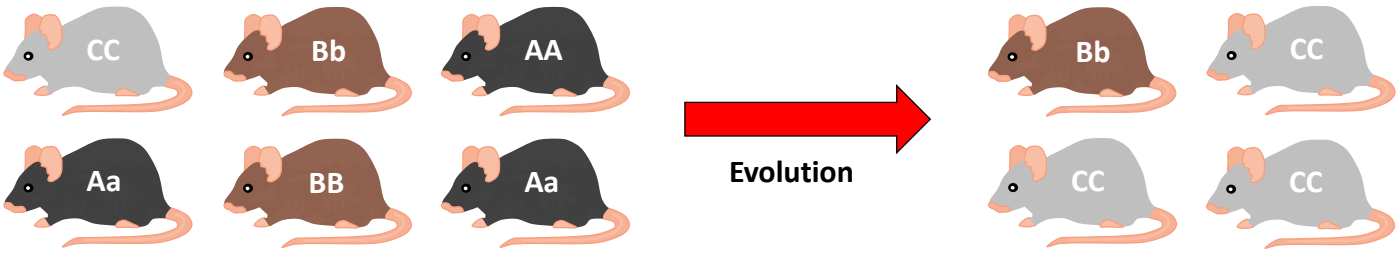
TOPIC: EVOLUTION

Key Knowledge:

- Causes of changing allele frequencies in a population's gene pool, including environmental selection pressures, genetic drift and gene flow; and mutations as the source of new alleles
- Biological consequences of changing allele frequencies in terms of increased and decreased genetic diversity
- Manipulation of gene pools through selective breeding programs

GENE POOLS

A gene pool represents the sum total of alleles for all genes present in a sexually reproducing population. A larger gene pool indicates higher levels of genetic diversity, which increases chances of species survival. New alleles can be created via **mutation** to increase the genetic diversity within a gene pool. These alleles can be either introduced or removed from a gene pool via **gene flow** (the movement of alleles into, or out of, a population as a result of immigration or emigration). Evolution describes the change in the frequency of specific alleles within a given population's gene pool. Evolutionary change can be driven by random events (**genetic drift**), environmental pressures (**natural selection**) or human direction (**artificial selection**). Evolution will reduce genetic diversity within a population as certain traits become more, or less, common.



Large gene pool = more biodiversity

Smaller gene pool

GENETIC VARIATION

Evolution involves cumulative changes in the **heritable** characteristics of a population (across generations). For evolution to occur, there must be variation among members of a species to allow for the differential selection of heritable traits. There are numerous mechanisms by which genetic variation can be achieved:

- **Mutations:** Changing the nucleotide sequence of a gene may change the associated phenotypic trait
- **Sexual reproduction:** Meiosis and fertilisation promotes the combination of genes from two sources
- **Chromosomal abnormalities:** Meiotic errors produce cells that have incorrect chromosome numbers

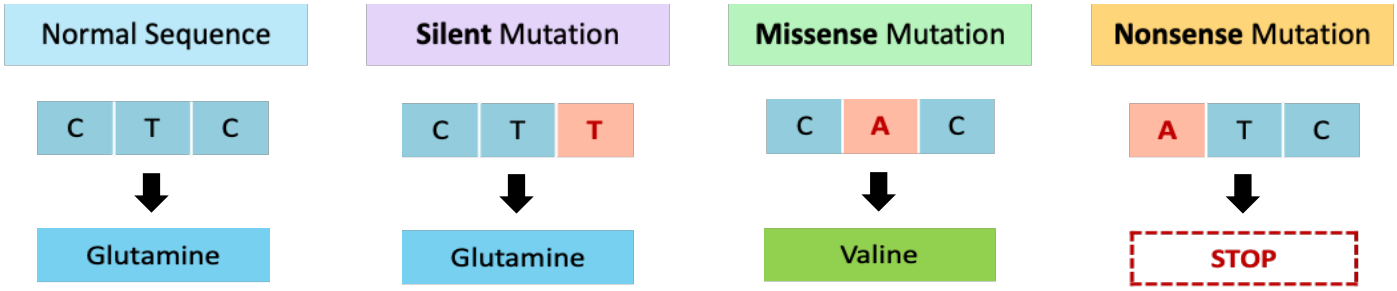
GENE MUTATIONS

A gene mutation is a change in the nucleotide sequence of a section of DNA encoding for a specific trait. Mutations can give rise to new versions of a gene (called alleles) and hence change the observable features of an organism (creating variation). Only **germ line** mutations (in gametes) will produce *heritable* variation, **somatic** mutations (in body cells) *cannot* be passed on to offspring. There are several types of mutations. Mutations that occur within non-coding DNA sequences will not typically change the characteristics of an organism (unless they impact gene expression levels) and hence do not usually influence genetic diversity.

1. POINT MUTATIONS

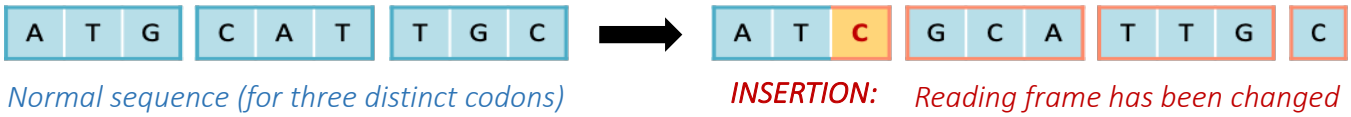
Point mutations involve a change to a single base within the DNA code. This may involve **base substitution** (one nucleotide is replaced by another) or **inversion** (two adjacent nucleotides swap positions). There are three main mechanisms by which a point mutation may affect polypeptide production by a specific gene:

- **Silent mutation:** When a DNA change does not alter amino acid sequence (due to codon degeneracy)
- **Missense mutation:** When a DNA change alters a single amino acid in the polypeptide chain
- **Nonsense mutation:** When a DNA change creates a premature STOP codon (truncating a polypeptide)



2. FRAMESHIFT MUTATIONS

Frameshift mutations involve a change that alters the reading frame of the DNA code. This may involve the addition (**insertion**) or removal (**deletion**) of a nucleotide, meaning every codon after this point is changed.



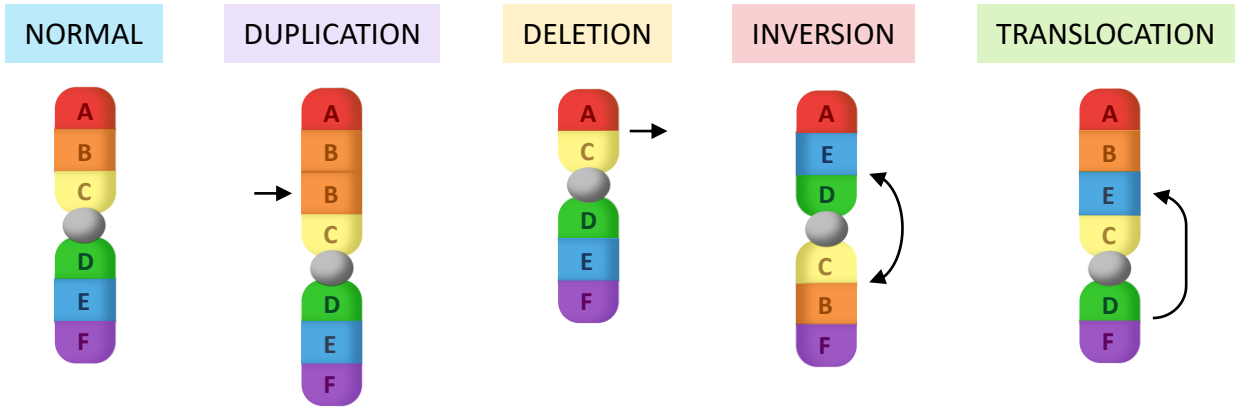
Normal sequence (for three distinct codons)

INSERTION: *Reading frame has been changed*

3. BLOCK MUTATIONS

Block mutations are changes to segments of a chromosome, leading to large scale changes to the DNA of an organism. Several different types of block mutations can occur to alter the sequence of a chromosome:

- **Duplications:** Part of a chromosome is copied, resulting in duplicate sections (increases expression)
- **Deletions:** A portion of the chromosome is removed (along with any genes contained in the segment)
- **Inversions:** A chromosome segment is rearranged in the reverse order of the original sequence
- **Translocations:** A chromosome sequence is moved to a new location (even a different chromosome)



Mutations are the only means of creating new alleles. Sexual reproduction can cause genetic reassortment (creating new *combinations* of alleles) and chromosomal abnormalities such as aneuploidy and polyploidy can result in *more, or less, copies* of a particular allele, but only via gene mutations can entirely new alleles be created within gene pools (although new alleles can be *introduced* from other gene pools by gene flow).

GENETIC DRIFT

Genetic drift is the change in the composition of a gene pool as a result of **chance** or **random events**. It will occur faster and be more significant in smaller populations, where chance events have a bigger impact on the gene pool. Conversely, larger populations will be less affected by random events and maintain more stable allele frequencies with lower genetic drift. There are two mechanisms by which population sizes may be reduced in order to increase genetic drift: **population bottlenecks** and the **founder effect**.

FACTORS AFFECTING DRIFT

Genetic drift is greater in **small populations**. There are two ways by which population size can be reduced:

Population Bottlenecks:

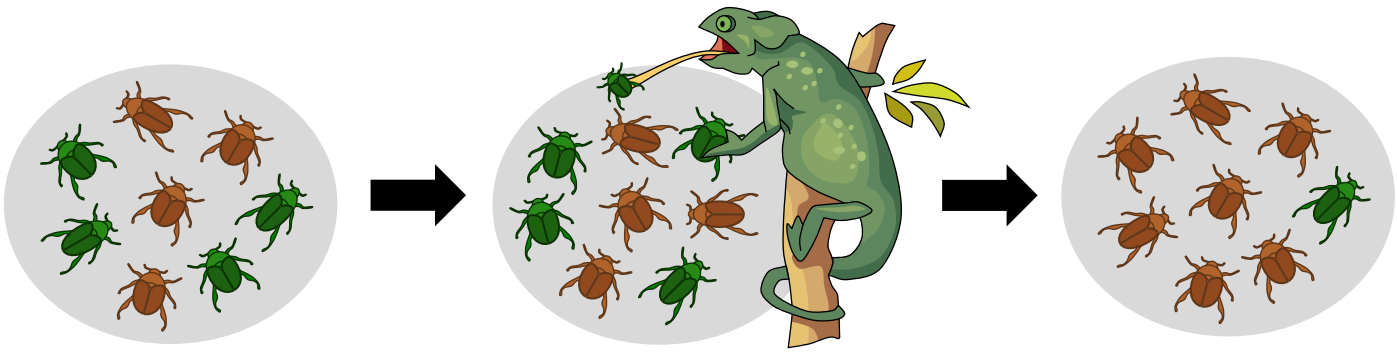
- Occur when chance events (forest fire, overhunting) reduce population size by an order of magnitude
- The original population is **not** preserved (it is replaced)

Founder Effect:

- Occur when a small group (subset) breaks away from a larger population to colonise a new territory
- The original population will still remain largely intact

NATURAL SELECTION

Natural selection describes the change in gene pool composition as a result of environmental **selection pressures**. Species will possess characteristics (called **adaptations**) that make individuals suited to the environment and way of life. Individuals with beneficial adaptations will be more likely to survive and reproduce ('survival of the fittest'), hence increasing the frequency of these inheritable characteristics in future generations. Adaptations that are detrimental to survival will become less common in a population as individuals are less likely to reproduce and pass on these features to offspring. Environmental conditions (selection pressures) will determine whether a particular adaptation is beneficial or detrimental to survival. Selection pressures include predators, competition, pathogens, access to resources and climate conditions.



SURVIVAL OF THE FITTEST

Natural selection requires a number of key conditions in order to occur:

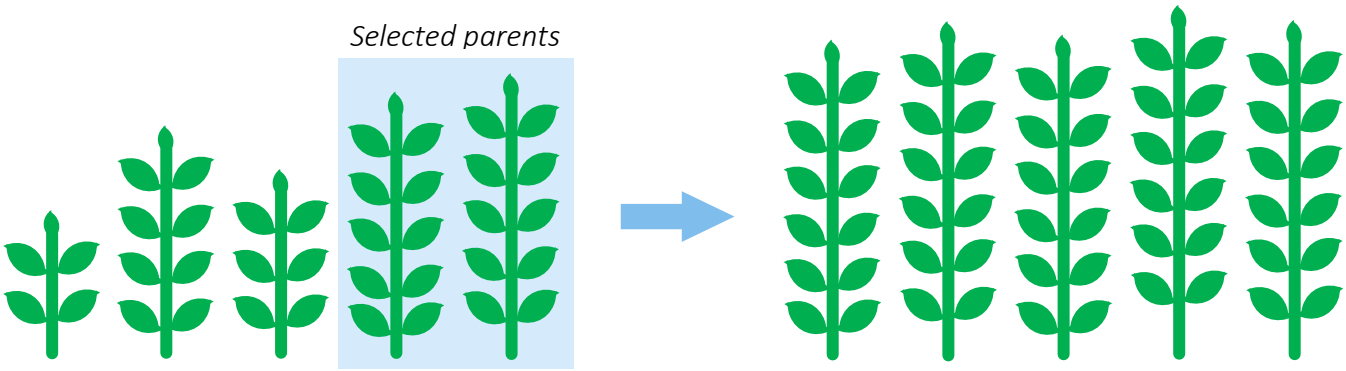
- **Inherited variation** must exist within the population (via mutations, etc.)
- **Competition** for survival results from an overproduction of offspring
- **Environmental selection pressures** lead to differential reproduction
- **Adaptations** which benefit survival will be passed on to offspring
- **Gene pool composition** changes (specifically: change in allele frequency)
- **Evolution** occurs within the population (cumulative change in traits)

Hint: ICE AGE

Natural selection changes the frequency of characteristics, leading to changes within a species. Biodiversity is reduced as the frequency of individuals with beneficial adaptations is increased within the population.

ARTIFICIAL SELECTION

Artificial selection occurs when man intervenes in the reproduction of a species to produce desired traits in offspring. This intervention can occur between generations (via **selective breeding**) or within a generation (via **genetic modification**). The systematic selection and propagation of phenotypic extremes will result in the rapid development of pronounced variability between the domesticated species and wild counterparts.



Phenotypic extremes selected and bred

Offspring present desired trait (height)

There are numerous examples of the manipulation of gene pools through selective breeding programmes:

- Plants of the genus *Brassica* have been bred to produce different foods by targeting plant components, including flower buds (broccoli or cauliflower), leaf buds (cabbage) and leaves (kale)
- Dog breeds show enormous variation due to the targeted selection of certain traits, including speed (greyhounds), intelligence (sheep dogs), size (chihuahuas) and aggression (mastiffs)

Scientists can also introduce desired traits into a species by genetically modifying the target organism. This may involve the transfer of genetic material from an alternate source via a delivery vehicle called a **vector**. A vector is a DNA molecule that is used as a vehicle to carry a gene of interest into a foreign cell. Bacterial plasmids are commonly used as vectors because they are capable of autonomous self-replication and expression. Introducing new alleles this way is called horizontal gene transfer (because the genes are transferred between organisms and not between generations – as is what occurs with selective breeding).