MUTATIONS

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D1.3.3 Consequences of insertions and deletions

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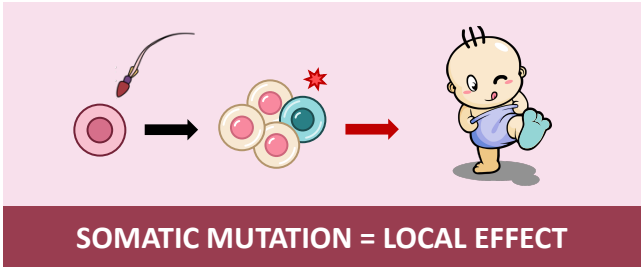
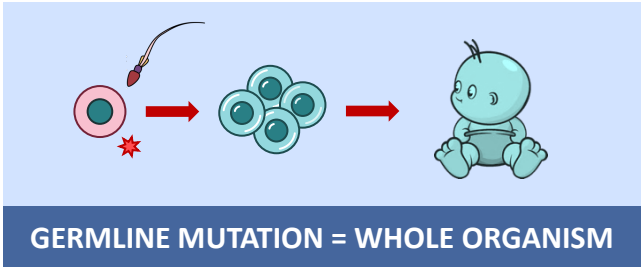
GENE MUTATIONS

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.



SOMATIC MUTATION = LOCAL EFFECT

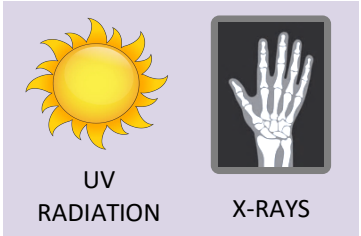


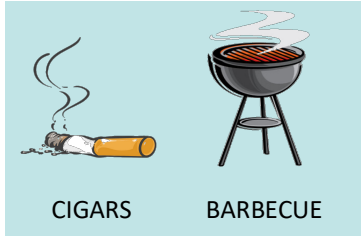
GERMLINE MUTATION = WHOLE ORGANISM

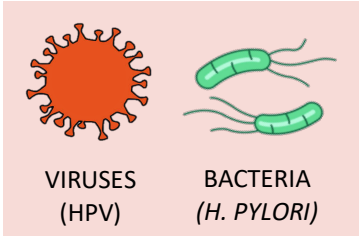
SOURCES OF MUTATION

Gene mutations can be caused by **proofreading errors** during DNA replication. Normally, the enzyme that is responsible for copying the DNA sequence (DNA polymerase) will detect and remove any incorrectly paired nucleotides. A mutation will result if an incorrectly incorporated nucleotide is not replaced when copied.

Gene mutations can also be caused by **mutagenic agents**. Mutagens induce a permanent change to the genetic material of an organism (increasing the frequency of mutations above a natural background level). *Physical mutagens* include certain forms of radiation (including X-rays and UV light). *Chemical mutagens* include substances such as reactive oxygen species, certain metals (e.g. arsenic) and alkylating agent (which can be formed by grilling meat). *Biological mutagens* include certain viruses (HPV) and bacteria *(H. pylori).*







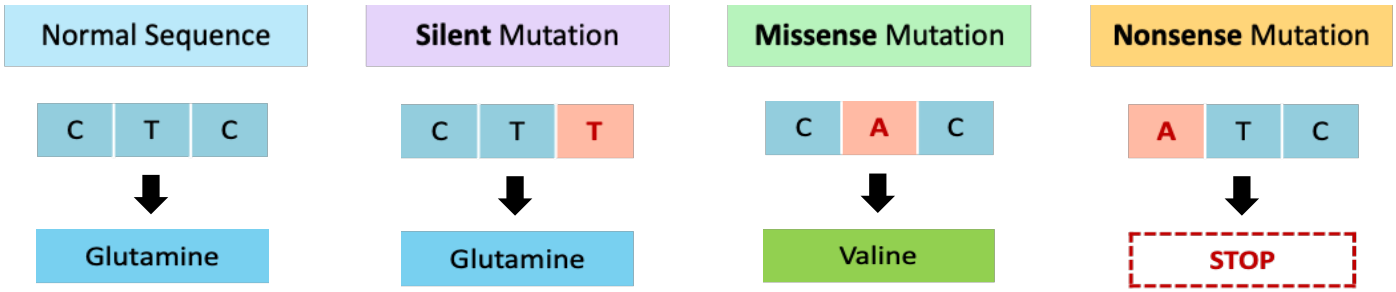
KEY:

- Physical
- Chemical
- Biological

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)



FRAMESHIFT MUTATIONS

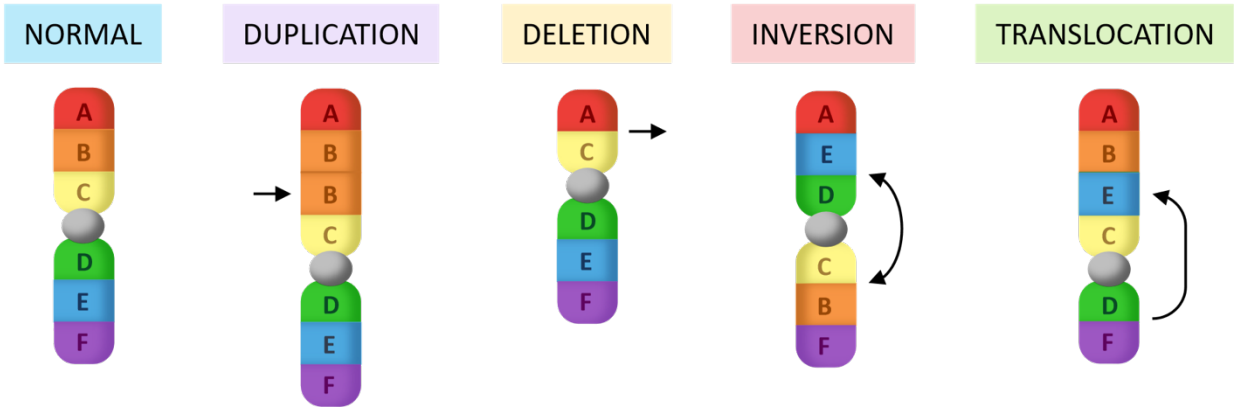
Frameshift mutations involve a change that alters the reading frame of the DNA code. This may involve the addition (**insertion**) or removal (**deletion**) of a nucleotide, meaning every codon after this point is changed.

Normal sequence (for three distinct codons) *INSERTION: Reading frame has been changed*

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 a population (but new alleles can be *introduced* from other populations via migration).