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Genotype and phenotype

Types of variants

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Family history and pedigree analysis

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Types of variants

Classification based on cell type:

Germline variants are variants in egg or sperm cells, and are able to be passed from parent to offspring.

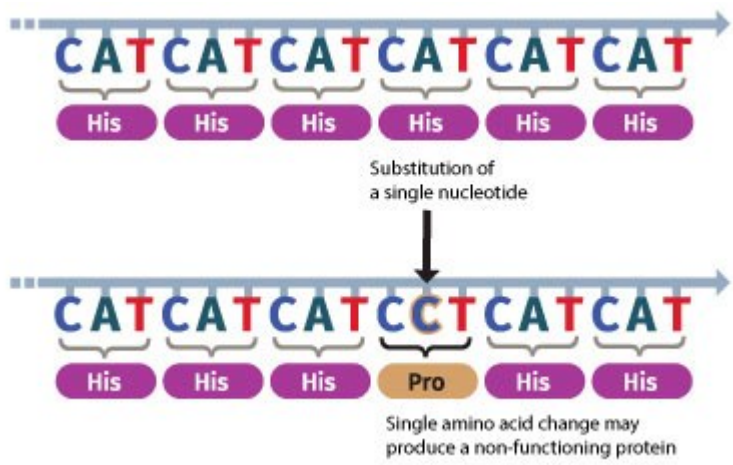
Somatic variants are present only in specific cells and are generally not hereditary. Somatic variants are acquired at some point during a person's lifetime, often due to environmental factors or errors in cell division. Some somatic variants will lead to the development of diseases such as cancer. Both germline and somatic variants have been implicated in many cancers.

Classification based on type of alteration:

Single nucleotide variant (SNV)

This is a substitution of a single nucleotide for another. A SNV can be rare in one population but common in a different population.

Sometimes SNVs are known as single nucleotide polymorphisms (SNPs), although SNV and SNPs are not interchangeable. To qualify as a SNP, the variant must be present in at least 1% of the population.



If a SNV occurs in a protein coding region, then this could result in either a

- A nucleotide substitution that does not result in a change in amino acid **(synonymous change)**. This is possible because multiple codons (sets of three nucleotides) code for the same amino acid.
- A nucleotide substitution leads to an amino acid substitution. This may or may not result in a pathogenic variant depending on the effect of the amino acid substitution on protein function and structure. This is called a **nonsynonymous change** or a missense variant (mutation).
- A nucleotide substitution results in a stop codon and consequently premature truncation of the protein. This protein is generally non-functional. This is known as a **stop gain change** or a nonsense variant (mutation).

Indel

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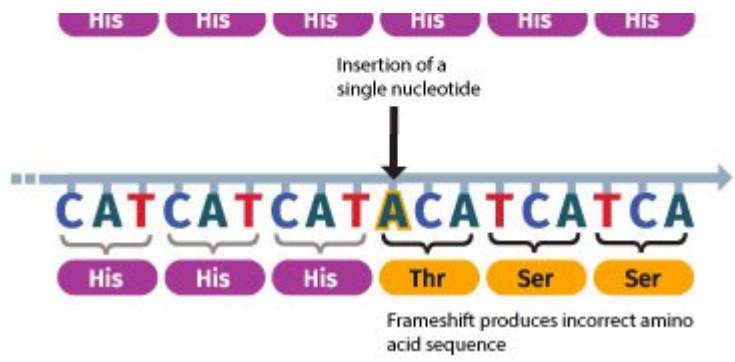
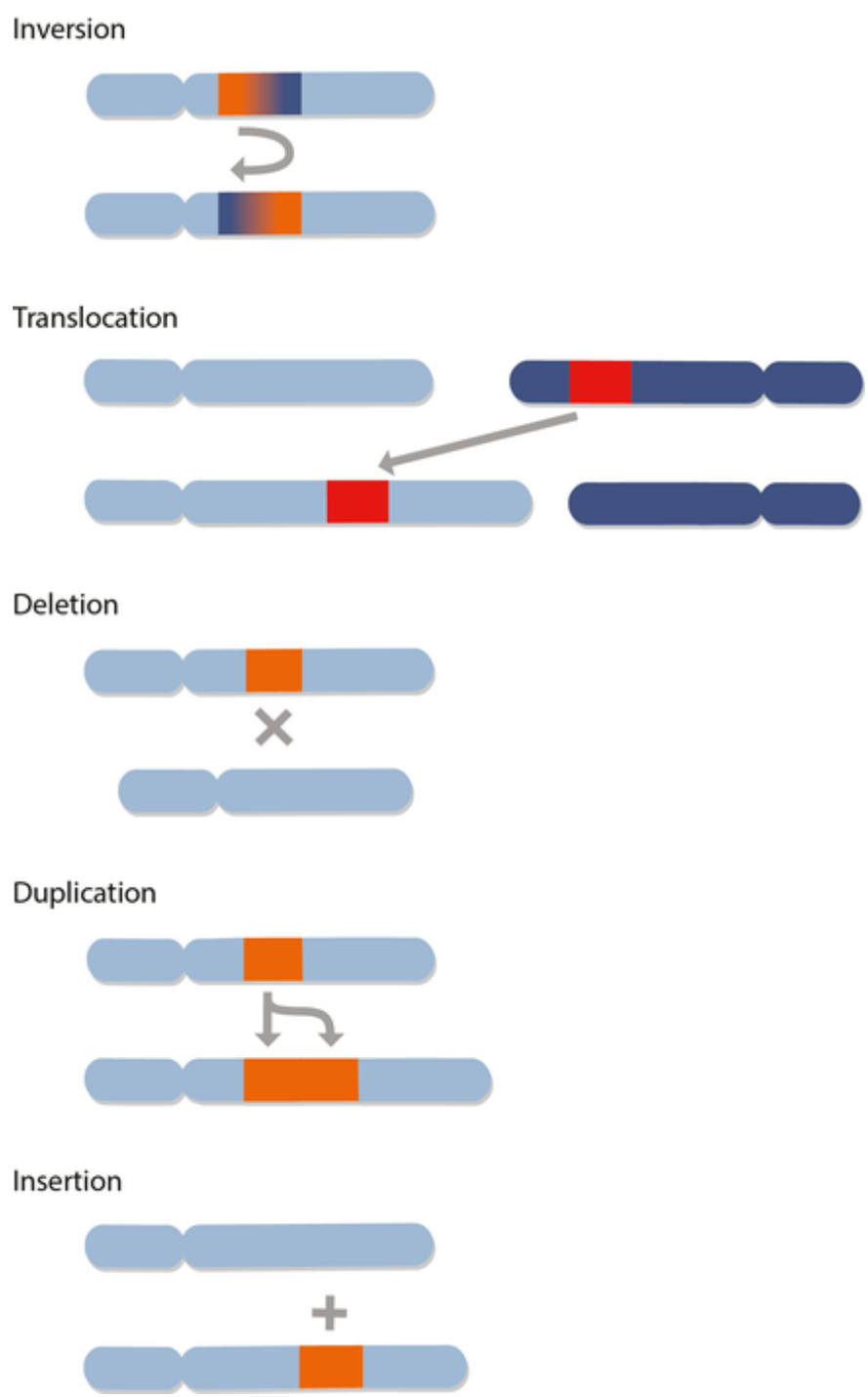


During translation, the mRNA sequence is read in groups of three bases (codons), with each codon corresponding to a particular amino acid. If the length of an indel is not a multiple of three, then it will likely result in a **frameshift.** A frameshift changes the reading frame of all the remaining bases resulting in the rest of the gene being incorrectly translated. This will drastically alter the product of translation, often leading to targeted decay of the mRNA .

Structural variation

This is a large (greater than 50 base pairs) rearrangement of part of the genome, and can be a deletion, duplication, insertion, inversion, translocation or often a combination of these. A **copy number variation (CNV)** is a duplication or deletion that changes the number of copies of a particular DNA segment within the genome. Structural variations have been implicated in a number of conditions, including polycystic kidney disease, cardiomyopathies, amyotrophic lateral sclerosis (ALS) and some cases of intellectual disability.



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Previous | Next: Inheritance patterns

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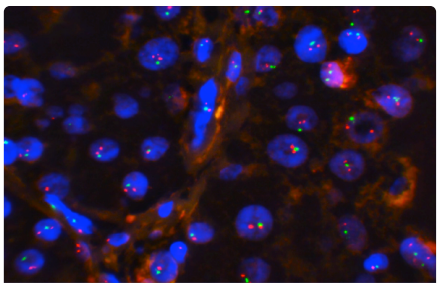

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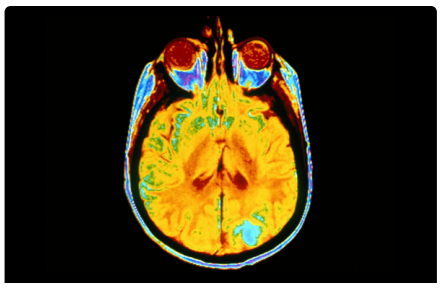

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