

Mutation and gene editing

Revision notes · IB Biology D1.3: substitutions, insertions and deletions, causes of mutation, and germ and somatic cells.

For IB Diploma Biology SL undefined. **HL only** is additional higher level content, for HL students only.

Mutation and gene editing **Spec D1.3**

Substitutions, insertions and deletions **Spec D1.3.1, D1.3.2, D1.3.3**

A **gene mutation** is a structural change to a gene at the molecular level: a change to its base sequence. There are three types.

Types of gene mutation		
Type	Change	Example (coding strand)
Substitution	One base is replaced by a different base.	GAG → GTG
Insertion	One or more extra nucleotides are added.	GAG CTA → GAA GCT A...
Deletion	One or more nucleotides are lost.	GAG CTA → GGC TA...

Consequences of base substitutions

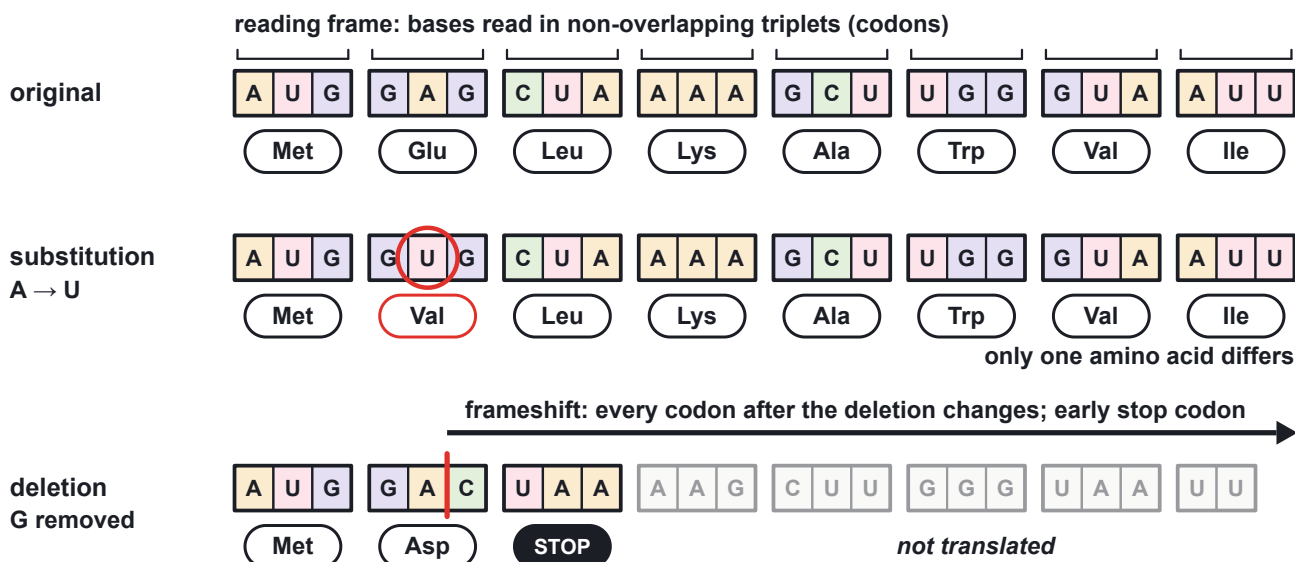
Single-nucleotide polymorphisms (SNPs) are positions in the genome where a single base differs between individuals. They are the result of base substitution mutations. A substitution changes only one codon, and because of the **degeneracy of the genetic code** it may or may not change a single amino acid:

- **Same amino acid:** the new codon codes for the same amino acid (for example GAA → GAG, both glutamic acid). The polypeptide is unchanged.
- **Different amino acid:** one amino acid in the polypeptide is changed (for example GAG → GUG, glutamic acid → valine). The effect may be small or, as in sickle-cell anaemia, large.
- **Stop codon:** the codon becomes a stop codon (for example UAC → UAA), so the polypeptide is cut short and is usually non-functional.

Consequences of insertions and deletions

Because mRNA is read in non-overlapping triplets, inserting or deleting one or two nucleotides shifts the reading frame. This **frameshift** changes every codon after the mutation, so the amino acid sequence from that point is altered and a premature stop codon often appears. The polypeptide is very likely to cease to function.

Inserting or deleting a multiple of three nucleotides does not cause a frameshift, but adds or removes amino acids. Major insertions or deletions of many nucleotides also make it likely that the polypeptide stops functioning.



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A substitution changes one codon at most; a deletion causes a **frameshift** that changes every codon after it.

Exam tip: Linking question: how does variation in subunit composition of polymers contribute to function? A single changed nucleotide in DNA can change one amino acid in a protein and alter its function.

Causes of mutation and randomness Spec D1.3.4, D1.3.5

Causes of gene mutation

A **mutagen** is an agent that increases the rate of mutation.

Causes of gene mutation		
Cause	Examples	How it causes mutation
Chemical mutagens	Benzo[a]pyrene and nitrosamines in tobacco smoke; mustard gas	React with bases or bind to DNA, causing bases to pair incorrectly during replication.
Mutagenic radiation	Ultraviolet (UV) light, X-rays, gamma rays, alpha particles from radioactive isotopes such as radon	UV causes adjacent pyrimidine bases (such as two thymines) to bond to each other; high-energy radiation ionises molecules and can break DNA strands.
Errors in DNA replication	A mismatched nucleotide not corrected by proofreading	A wrong base is copied into the new strand and then into every later copy.
Errors in DNA repair	Damaged DNA repaired incorrectly	The repaired region has a changed base sequence.

Randomness in mutation

Mutations are **random**. They can occur anywhere in the base sequences of a genome, although some bases have a higher probability of mutating than others (for example, a cytosine with a methyl group added, often where C is followed by G, can lose an amino group and become thymine, so C → T substitutions are especially common at these sites). There is **no known natural mechanism for making a deliberate change to a particular base** with the purpose of changing a trait. An organism cannot produce a mutation because it would be useful.

Common mistake: Do not write that organisms mutate in order to adapt. Mutations happen at random; natural selection then acts on the variation they produce.

Germ and somatic cells, and mutation as a source of variation Spec D1.3.6, D1.3.7

Germ cells and somatic cells

Consequences of mutation in different cells		
	Germ cells	Somatic cells
What they are	Cells that produce gametes (and the gametes themselves)	All other body cells
Inherited?	Yes: a mutated gene can be passed to offspring	No: not passed to offspring
Cells affected	Every cell of an offspring that inherits it	Only the mutated cell and the cells produced from it by mitosis
Possible consequence	Inherited genetic disease, or new variation in the population	Cancer , if mutations affect genes that control cell division

Cancer develops when mutations in a somatic cell cause it to divide repeatedly without control, forming a tumour. Usually several mutations in the same cell line are needed, which is one reason cancer becomes more common with age and with exposure to mutagens.

Mutation as a source of genetic variation

Gene mutation is the **original source of all genetic variation**: it is the only process that creates new alleles. Meiosis and fertilisation shuffle existing alleles into new combinations, but cannot create new ones. Most mutations are either harmful or neutral for an individual organism, but in a species over the long term they are **essential for evolution by natural selection**, because selection can only act on variation that already exists.

Exam tip: Nature of science: commercial genetic tests can give information about potential future health and disease risk. Without expert interpretation, this information could be misunderstood and cause needless worry or false reassurance.

Exam tip: Linking question: how can natural selection lead to both a reduction in variation and an increase in biological diversity? Mutation supplies the variation; selection removes some alleles within a population but can drive populations apart into new species.

Quick check

1. What is a single-nucleotide polymorphism (SNP)?
2. Why does deleting one base usually have a larger effect than substituting one base?
3. Give one chemical mutagen and one form of mutagenic radiation.
4. Why are somatic mutations not inherited?

Quick check answers

Mutation and gene editing

- 1 . A position in the genome where a single base differs between individuals, resulting from a base substitution mutation.
- 2 . It shifts the reading frame, changing every codon after the mutation.
- 3 . Benzo[a]pyrene (in tobacco smoke); ultraviolet light (or X-rays, gamma rays).
- 4 . They are not in germ cells, so they are not passed into gametes.