Knowledge organiser for KS5 Biology Alterations of the sequence of bases in DNA can alter the structure of proteins at Saint Ambrose College 2020+

What is covered in this unit?

Gene mutations; Mutagenic agents; Different amino acid sequence in encoded polypeptide

Key vocabulary

Gene mutation – any change to one or more nucleotide bases, or any rearrangement of bases, in DNA

Mutagenic agents – basic mutation rate increased by outside factors

Key facts-

Substitution of bases

The type of gene mutation in which a nucleotide in a section of a DNA molecule is replaced by another nucleotide that has a different base is known as a substitution. Depending on which new base is substituted for the original base, there are three possible consequences:

- The formation of one of the three stop codons that mark the end of a polypeptide chain. As a result the production of the polypeptide coded for by the section of DNA would be stopped prematurely. The final protein would almost certainly be significantly different and the protein could not perform its normal function.
- The formation of a codon for a different amino acid, meaning that the structure of the polypeptide produced would differ in a single amino acid. The protein of which this polypeptide is a part may differ in shape and not function properly. For example, if it is an enzyme, its active site may no longer fit the substrate and it will not catalyse the reaction. An example of this form of substitution mutation causes a condition called sickle cell anaemia.
- The formation of a different codon but one that produces a codon for the same amino acid as before. This is because the genetic code is degenerate and so most amino acids have more than one codon. The mutation therefore has no effect on the polypeptide produced and so the mutation will have no effect.

Other types of gene mutation

There are a number of other ways in which the base sequence of DNA may be changed. These include:

- **Addition of bases** – an extra base becomes inserted in the sequence. This usually has a similar effect to a base deletion in that there is usually a frame shift and the whole sequence of triplets becomes altered. The frame shift is to the right not to the left as it is when a base is deleted. If three extra bases are added, or any multiple of three bases, there will not be a frame shift. The resulting polypeptide will be different from the one produced from a non-mutant gene, but not to the same extent as if there was a frame shift.
- **Duplication of bases** – one or more bases are repeated. This produces a frame shift to the right.
- **Inversion of bases** – a group of bases become separated from the DNA sequence and rejoin at the same position but in the inverse order (back to front). The base sequence of this portion is therefore reversed and effects the amino acid sequence that results.
- **Translocation of bases** – a group of bases become separated from the DNA sequence on one chromosome and become inserted into the DNA sequence of a different chromosome. Translocations often have significant effects on gene expression leading to an abnormal phenotype. These effects include the development of certain forms of cancer and also reduced fertility.

Mutations have both costs and benefits. On the one hand they produce the genetic diversity necessary for natural selection and speciation (see Topics 18.4 and 18.5). On the other hand they are almost always harmful and produce an organism that is less well suited to its environment. Additionally, mutations that occur in body cells rather than in gametes leading to disruption of normal cellular activities, such as cell division, for example, cancer.

Possible homework tasks | **Stretch & challenge (wider reading/independent work)**

H/W:including project, Kerboodle, Kahoot.

Stretch: modelling tasks, Biological Science Review research.