

**Higher Human Biology**

**Human cells: Mutations (Key Area 4)**

By the end of this topic I will be able to:

1. State what is meant by a mutation and the effects they can have on a protein.
2. State what a single gene mutation is and what causes them.
3. To understand that missense, nonsense and splice-site mutations are all caused by nucleotide substitutions.
4. To state what is meant by missense, nonsense and splice-site mutations and describe their effects on the synthesis of proteins.
5. Describe the effects of an insertion or deletion as a frame-shift mutation.
6. State that mutations can occur in chromosomes which can be lethal.
7. Describe these chromosome mutations as duplication, deletion, inversion or translocation and explain the effects of these on chromosome structure.