This changes the amino acid used to valine, and this simple change in the amino acid sequence leads to a significant change in the structure of the haemoglobin molecule synthesised, with devastating effects on health.

The consequences of other mutations such as small-scale deletions or insertions of one or more nucleotides depend on how the reading frame is altered. If any number of nucleotides other than exact multiples of three are inserted or deleted, the reading frame downstream of that segment is disrupted (frameshift mutation). Where exact multiples of three nucleotides are involved, there is less disruption and the outcome can result in milder disease (in-frame mutation). Duchenne muscular dystrophy results from a frameshift mutation (deletion) within the dystrophin gene. The milder Becker muscular dystrophy results from an in-frame deletion in the same gene.

Mis-sense, nonsense, frameshift and in-frame mutations can all occur in the same gene. The cystic fibrosis transmembrane regulator (CFTR) gene on chromosome 7 is an example of this (Collins, 2009). Where both chromosomes of a pair carry a mutation in the CFTR gene, the individual will have cystic fibrosis, although the severity and clinical outcome will vary according to the type of mutation.

Another mutation that can be associated with disease is where a short sequence of nucleotides (typically three) is repeated within the gene, and the number of repeats increases above a specific threshold. Huntington’s disease is caused by repeating units of CAG within the HD gene on chromosome 4. The normal range of such repeats is 5-35, but anything above 37 is pathogenic, leading to the disease. The size of the expanding repeat can vary substantially in other genes too, up to many thousands, and repeat length is often correlated with the severity and/or age of onset of the condition. Conditions caused by expanding repeats include Fragile X syndrome and myotonic dystrophy.

Deletions, insertions and inversions of genetic material are all alterations to the chromosomal structure. These changes can involve much larger segments of DNA, sometimes spanning one or more genes, where there have been breaks in the DNA molecule that have not been repaired correctly. Alterations to chromosome number as a result of faults during meiosis also lead to duplication or deletion of genetic material, such as with Down’s syndrome.

The diseases that can result from mutations within a single gene show characteristic patterns of inheritance, as first described by Mendel. The type of pattern depends on the outcome of the mutation, that is, whether there is a loss or gain of function. If the outcome is sufficiently marked for it to be shown when only one gene of a pair is mutated, the inheritance pattern is said to be dominant. There is a