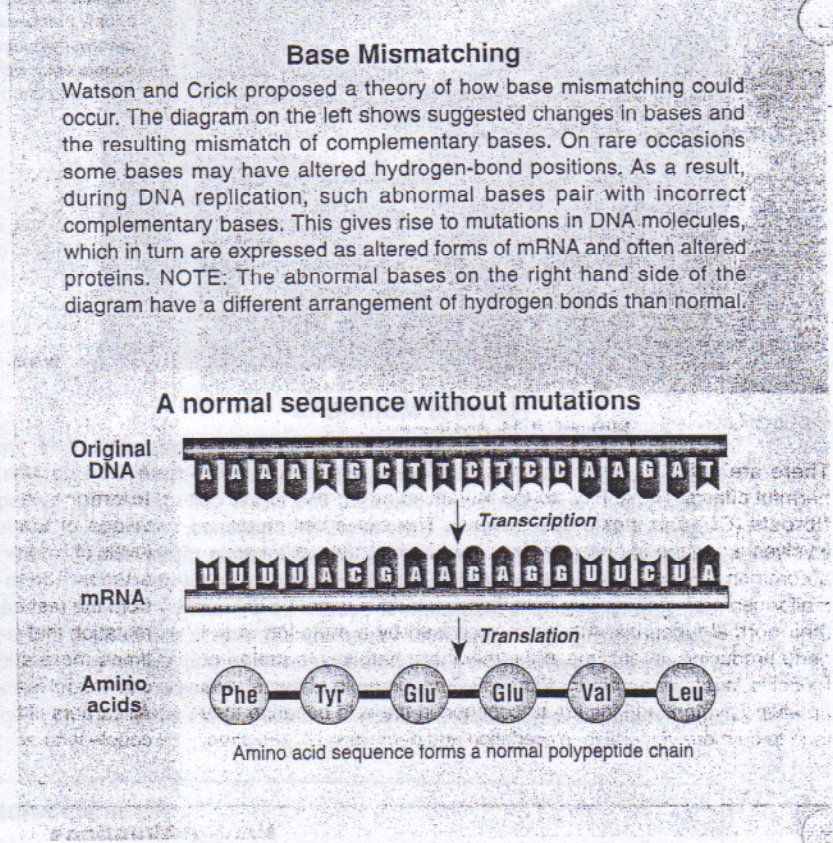
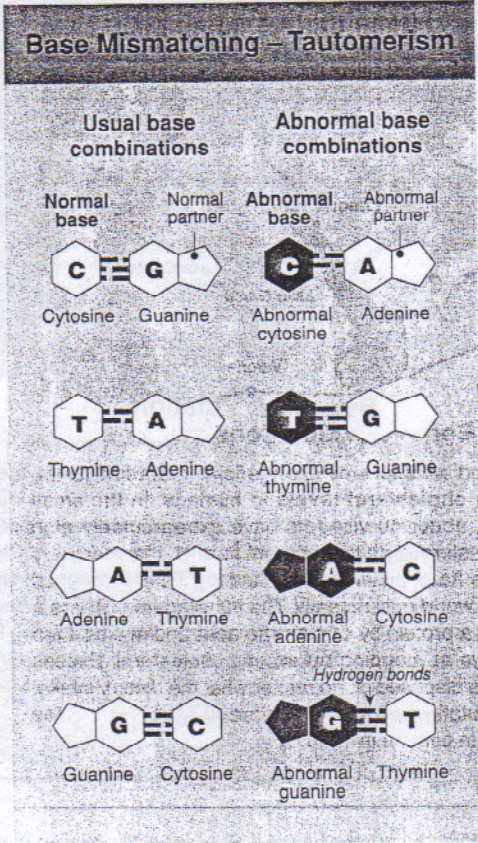


Gene Mutations

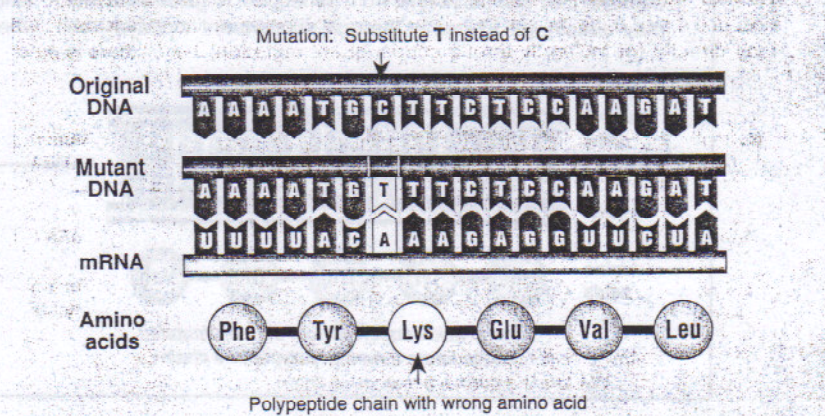
Gene mutations are small, localised changes in the structure of a DNA strand. These mutations may involve change in a single nucleotide (often called **point mutations**), or they may involve changes to a triplet (e.g. deletion or triplet repeat). If one amino acid in a protein is wrong, the biological function of the entire protein can be disrupted. Not all mutations may result in altered proteins. Because of the degeneracy of the genetic code, a

substitution of the 3rd base in a codon may code for the same amino acid. The diagrams below and opposite show how various point mutations can occur. These alterations in the DNA are at the **nucleotide** level where individual **codons** are affected. Alteration of the precise nucleotide sequence of a coded gene in turn alters the mRNA transcribed from the mutated DNA and may affect the polypeptide chain that it normally creates.



Missense substitution

A single base is substituted for another base which may result in a codon that codes for a different amino acid. Some substitutions, however, may still code for the same amino acid, because of the high degree of degeneracy in the genetic code (i.e. many amino acids have 4 or 6 codons coding for them). In the illustrated example, placing a T where a C should have been, results in the amino acid **lysine** appearing where **glutamic acid** should be. This could affect how this protein functions.



Nonsense substitution

Some amino acids can be coded for by 4 or 6 different codons and are therefore less affected by substitutions. In the example illustrated, a single base substitution in the first nucleotide of the third codon has a dramatic effect on the nature of the polypeptide chain it is coding for. The codon no longer codes for an amino acid, but instead is an instruction for the termination of the translation process of protein synthesis. This results in a very short polypeptide chain that is likely to have little or no function since the **STOP** codon is introduced near the **START** codon.

