**Sickle Cell Anaemia**

*a disease caused by a genetic mutation*

*The difference in the adult haemoglobin of sickle cell anaemic sufferers and the rest of the population was discovered in 1949 by a team led by Linus Pauling. It was the first time that a disease was shown to be caused by a faulty molecule. Ingram in 1956 showed exactly what the difference was, and the entire amino acid sequence has since been determined.*

Haemoglobin is a globular protein found in red blood cells.

It is made up of 4 polypeptide chains (2 x alpha chains and 2 x beta chains), surrounding an iron containing haem group.

The alpha chain is 141 amino acids long, while the beta chain is 146 amino acids long.

The mutation is found on chromosome 11 in the gene coding for the beta chain.

Normal beta chain gene: GTCCATCTGACTCCAGAGGAGAAG

SCA beta chain mutation: GTCCATCTGACTCCAGTGGAGAAG

1. *Use your codon table to work out the initial sequence of* ***amino acids*** *for a normal beta chain polypeptide (write it in the space below)*
2. *What* ***type*** *of mutation is found in the SCA beta chain DNA? Describe which codon is affected and exactly what has changed.*
3. *Write out the* ***mRNA*** *that will be formed when the mutated gene is transcribed.*
4. *What effect does that have on the* ***beta chain polypeptide*** *at translation? Describe exactly which amino acid(s) are affected and how.*