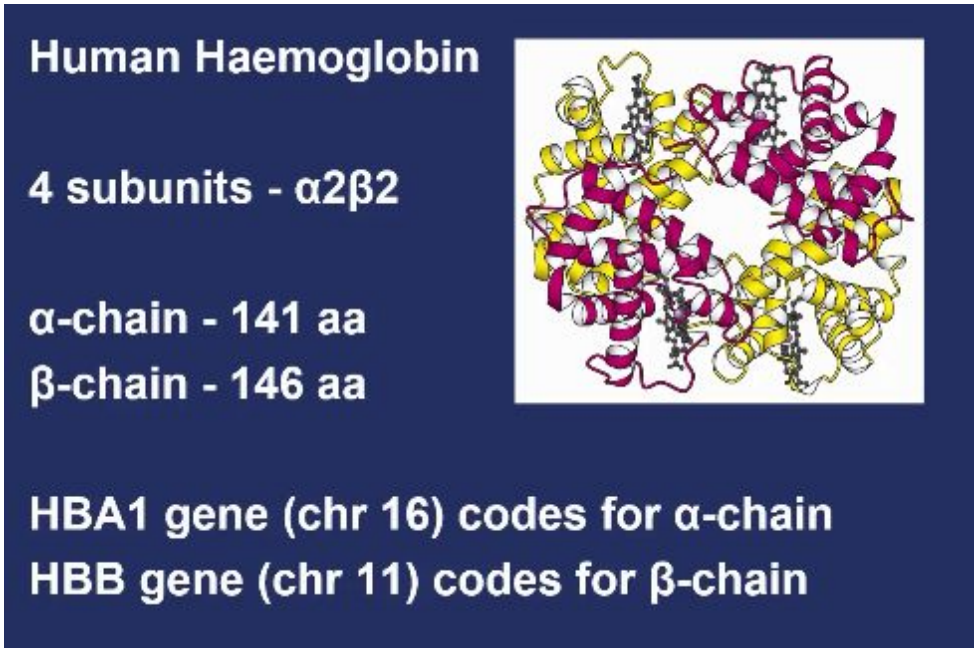


- the protein haemoglobin has 4 subunits in total, 2 alpha and 2 beta subunits.
- The alpha chain = 141 amino acids long
- The beta chain = 146 amino acids long
- Both of these proteins are coded by a gene.
- The HBA1 gene located on chromosome 16 codes for the alpha chain.
- The HBB gene located on chromosome 11 codes for the beta chain.

What is a gene?

- 1 gene = 1 functional unit
- This 'functional unit' can be a sequence of amino acids in a polypeptide Or… a sequence of nucleotides in an untranslated RNA

Where is a gene located?

- Genes have a chromosomal location

How many?

- Humans have around 25,000 genes
- Fruit flies have around 15,000 genes
- E.coli has around 4,500 genes.

The amount of genes is not important, the regulation of their expression is.

- In sickle cell anaemia the 7th codon is 5' GTG producing Val instead of Glu

Section 9: Phenotype and Genotype:

Phenotype:

- The form (trait) you will see.
- Genotype:
- The genetic makeup.

Section 10: Human haemoglobin mutations:

- Mutations are common all over the code
- Missense mutations are when a single letter is changed
- The 7th codon may have many missense mutations which changes the amino acid produced:

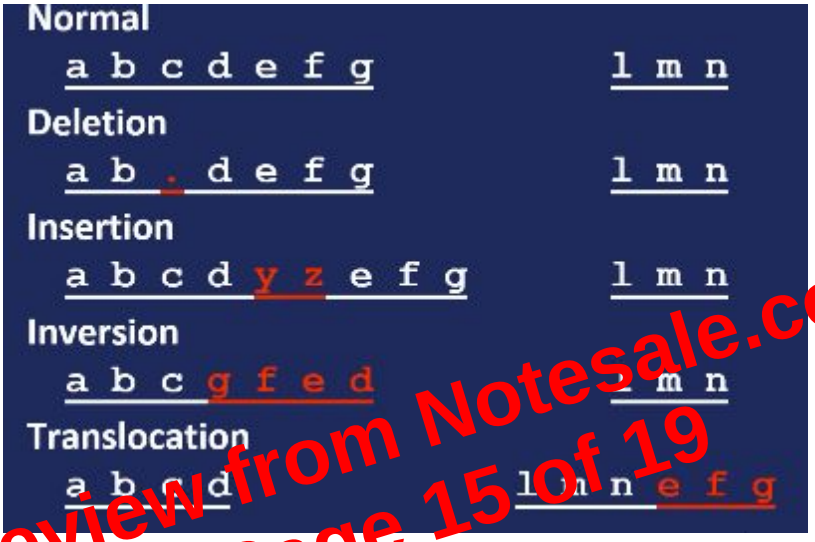
aa change	Name of Hb	Codon 7 change
Glu ⇨ Val	S	GAG ⇨ GTG *(Missense)*
Glu ⇨ Lys	C	GAG ⇨ AAG *(Missense)*
Glu ⇨ Ala	G Makassar	GAG ⇨ GCG *(Missense)*
Loss of Glu	Leiden	*Deletion* of GAG
NB. Glu ⇨ Glu i.e. NO change	normal	GAG ⇨ GAA *(Neutral, Silent)*

Sickle cell anaemia

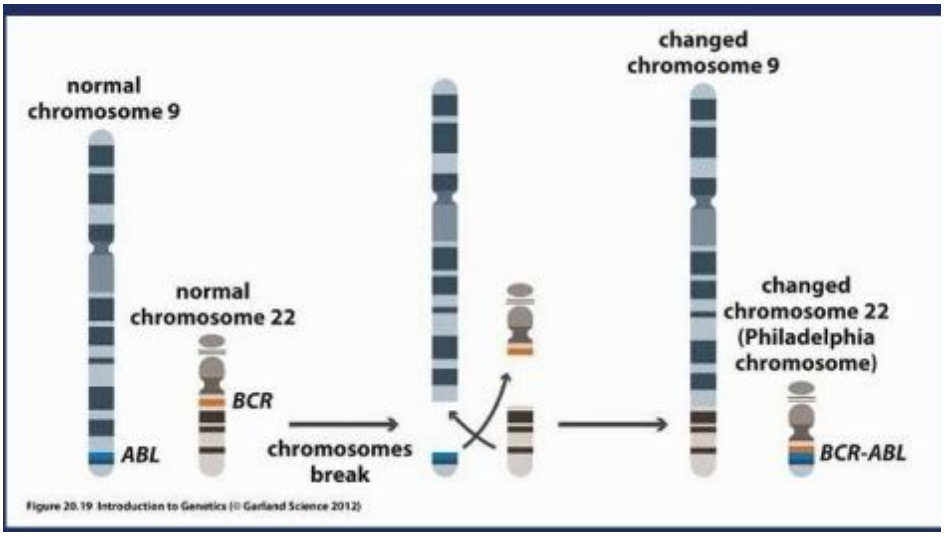
Deletion of the entire amino acid

- Single base changes don't necessarily have the same phenotype

- **Insertion**: A type of mutation characterized by the insertion of one or few nucleotide base pairs to a chromosome.
- **Deletion:** a part of a chromosome or a sequence of DNA is lost during DNA replication.
- Any number of nucleotides can be deleted, from a single base to an entire piece of chromosome.
- **Inversion**: a mutation where parts of a chromosome are flipped or inverted.
- **Translocation**: Chromosomal translocation that is a chromosomal segment is moved from one position to another, either within the same chromosome or to another chromosome.



Transposable element insertions:



- In the breakage of chromosome 9 and 22, and the last bits of the Q arms can swap over