

TOPIC 3.1: GENES

Genes versus Alleles

A **gene** is a heritable factor that consists of a sequence of DNA and influences a specific trait

- The position of a gene on a chromosome is the **locus**

Alleles are the alternate forms of a gene that code for the different variations of a specific trait

- Alleles for a specific gene will differ by only a few bases

New alleles may be formed as a result of gene mutations

Genome

A **genome** describes the totality of the genetic information in an organism

- It includes all genes and non-coding sequences

The Human Genome Project was completed in 2003 and mapped the entire base sequence of human genes

- Human cells typically have 46 chromosomes
- The human genome consists of ~3 billion base pairs
- It contains roughly 21,000 genes (although estimates vary)

The genomes of other organisms are now being sequenced

Mutations

A **gene mutation** is a change in the base sequence of a section of DNA coding for a particular characteristic

- Gene mutations may be beneficial, detrimental or neutral

Gene mutations may be described as either:

- Somatic – Occurs in a body cell and affects a tissue
- Germline – Occurs in a gamete and affects offspring

Point mutations may include either:





- Substitutions (either silent, missense or nonsense)
- Frameshifts (insertions or deletions)

Mutations can arise spontaneously as copying errors during DNA replication or can be induced by mutagenic agents

Original Sequence		Point Mutation		
		Silent	Missense	Nonsense
DNA	TTC	TTT	TCC	ATC
RNA	AAG	AAA	AGG	UAG
Protein	Lys	Lys	Arg	STOP

Genetics Comparisons

There is no clear correlation between genetic complexity and chromosome numbers, genome size or the number of genes

Species	Diploid Number	Genome Size (Mb)	Gene Count	
Virus <i>T4 Phage</i>	n/a	0.17	280	
Bacteria <i>E. coli</i>	n/a	4.6	4200	
Fruit Fly <i>D. melanogaster</i>	8	130	13,000	
Roundworm <i>P. equorum</i>	4	185	14,000	
Rice <i>O. sativa</i>	24	470	38,000	
Canopy Plant <i>P. japonica</i>	40	150,000	?	
Dog <i>C. familiaris</i>	78	2,900	20,000	
Chimpanzee <i>P. troglodytes</i>	48	3,300	22,000	
Human <i>H. sapiens</i>	46	3,200	21,000	

Sickle Cell Anemia

Cause of Sickle Cell Anemia:

Base substitution: GAG → GUG (6th codon: hemoglobin beta)

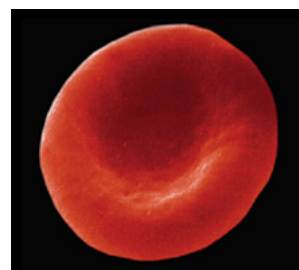
Amino acid change: Glutamic acid → Valine (Glu → Val)

Consequences of Sickle Cell Anemia:

- Alters haemoglobin structure (forms insoluble strands)
- Cannot transport oxygen effectively (causing fatigue)
- Red blood cells adopt a sickle shape (may form clots)
- Sickle cells are destroyed at a higher rate (causes anemia)

Heterozygous Advantage:

- Sickle cell anemia is a codominant trait and heterozygous individuals demonstrate an increased resistance to malaria



Normal Blood Cell



Sickle Blood Cell