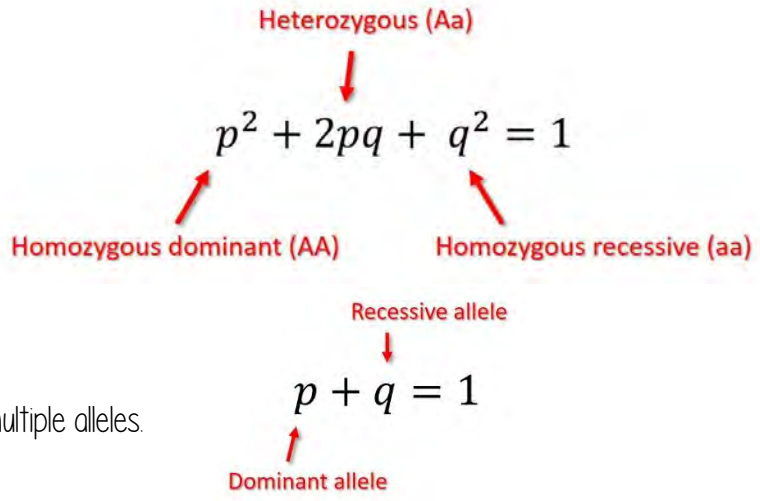
Genetics

Hardy-Weinberg Equilibrium

Evolution is a change in allele frequencies within a gene pool.

Evolution will not occur if:
> mutation is not occurring.
> natural selection is not occurring.
> the population is infinitely large.
> all members of the population breed.
> all mating is random.
> everyone produces the same number of offspring.
> there is no emigration or immigration.

A polygenic trait refers to a phenotype resulting from multiple alleles.



DNA Sequencing

DNA sequencing is the process of determining the order of nucleotides in a given DNA fragment.

Type II restriction endonucleases were first used for DNA sequencing; however, they only produced a limited amount of scattered data.

Maxam-Gilbert DNA sequencing chemically modifies then cleaves DNA.

Sanger Sequencing

A chain termination method of sequencing, also known as di-deoxy chain termination sequencing.

The technique involves sequence specific termination of DNA synthesis using modified nucleotide bases.

Sanger sequencing uses:
> template DNA: in-chain terminator sequencing DNA extension is initiated at a specific site on the template DNA.
> primer: must use a short oligonucleotide which is complementary to the template.
> DNA polymerase: the primer is extended using DNA polymerase.
> all four dNTPs.
> all four di-deoxynucleotide bases.

DNA polymerase catalyses the addition of the next dNTP to the 3' -OH group on the ribose of the previous nucleotide.

Di-deoxynucleotide base terminates elongation as it lacks a crucial 3'-OH group.

Next Generation Sequencing

Capable of sequencing whole genomes using small lengths of DNA reassembled with computer algorithms.

PCR

Used for amplifying a target sequence of DNA.

Requires:

> DNA template – extracted from sample, in solution, and non-specific.

Pre-Implantation Genetic Diagnosis

Uses *in vitro* fertilisation technology, with no need to terminate the pregnancy. However, this is a complicated procedure and only offers a 15-20% success rate.

Gene Therapy

Gene therapy refers to the introduction of nucleic acids into cells to correct or prevent pathologies. This can either occur *ex vivo* or *in vivo*. Vectors are used to transfer genes into cells.

Ideal Vectors

> can deliver a large amount of DNA.
> are limited to target cell type.
> are transcriptionally competent for the desired length of time.
> are easy to produce in high concentrations.
> are immunologically neutral.

Retroviral Vectors

+ Relatively safe
+ Integrates into the host genome.
- Only infects mitotic cells.
- Random integration that may disrupt the host genome.
- Difficult to produce on a large scale.

Adenoviral Vectors

Adenoviral vectors use DNA and replicate within the nucleus. These vectors are not integrated into the host genome.

+ Efficient transfection in non-dividing cells.
+ Easy to generate.
- Produces an immune response.
- Does not integrate so only provides a short-lived expression.

Non-Viral Vectors

Non-viral vectors inject naked plasmid DNA. They consist of a promoter region and the gene of interest. An example of a non-viral vector are liposomes. These interact with DNA, incorporate into the cell membrane, and release the DNA into the cytoplasm.

+ Easy to produce.
+ Can carry large amounts of DNA.
+ Targets specific cells.
+ Does not induce an immune response.
- Low transfection efficiency.
- Transient expression.
- Inhibited by blood serum.