POPULATION GENETICS

Content Statements:

D4.1.9	Concept of a gene pool
D4.1.10	Allele frequencies of geographically isolated populations
D4.1.11	Changes in allele frequency in the gene pool as a consequence of natural selection between individuals according to differences in their heritable traits
D4.1.12	Differences between directional, disruptive and stabilising selection
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D4.1.14	Hardy-Weinberg conditions that must be maintained for a population to be in equilibrium
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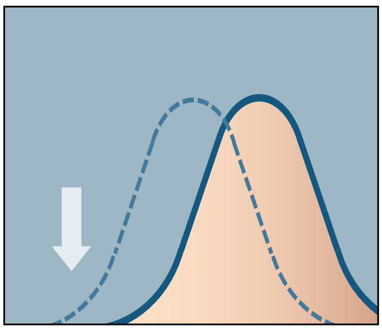
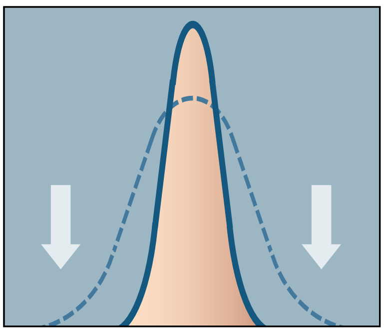
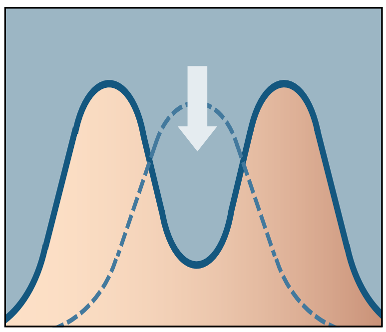
GENE POOLS

A gene pool represents the sum total of alleles for all genes present in a sexually reproducing population. A large gene pool represents high amounts of genetic diversity, increasing the chances of species survival. As each individual contains two alleles for each gene (diploid), the number of alleles in a gene pool should be roughly twice the population size. Gene pools can be used to determine **allele frequency** – the prevalence of a given allele for a specific gene. Allele frequencies may change in a population over time as a result of evolutionary processes (natural or artificial selection). Large and stable populations experience less change.

NATURAL SELECTION

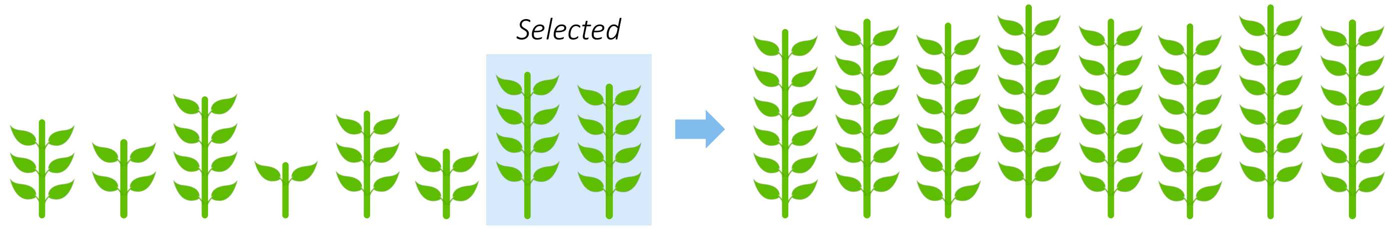
Charles Darwin proposed the theory of natural selection, whereby changes in characteristics in populations were driven by environmental selection pressures influencing survival and reproduction rates. These ideas have since been incorporated with a modern understanding of genetics (**Neo-Darwinism**). Evolution is now widely recognised to be a *change in the allele frequency of a gene pool* over many successive generations.

There are three different types of mechanisms by which allele frequency may change via natural selection:

Directional Selection	Stabilising Selection	Disruptive Selection
A phenotypic extreme selected at the expense of other extreme	Intermediate phenotype selected at the expense of both extremes	Phenotypic extremes favoured at the expense of the intermediates
Occurs in changing environment	Occurs in stable environments	Due to fluctuating conditions
Example: Antibiotic resistance	**Example:** Human birth weights	**Example:** Moth pigmentation
		

ARTIFICIAL SELECTION

Artificial selection involves the <u>deliberate</u> intervention by humans in the reproduction of species to produce desired traits in offspring. This can involve **selective breeding** or horizontal gene transfer (**transgenics**). The selection of phenotypic extremes will result in the rapid development of pronounced variation between the domesticated species and wild counterparts. <u>Unintended</u> human actions cause natural selection to occur.



Phenotypic extremes are selected and bred

Offspring present the desired trait (height)

There are numerous examples of the manipulation of gene pools through selective breeding programmes:

- Plants of the genus *Brassica* have been bred to produce different foods by targeting plant components, including flower buds (broccoli or cauliflower), leaf buds (cabbage or brussels sprouts) and leaves (kale)
- Dog breeds show enormous variability due to the targeted selection of certain traits – including speed (greyhounds), intelligence (sheep dogs), size (poodles), disposition (labradors) or aggression (mastiffs)

HARDY-WEINBERG

The Hardy-Weinberg equations function to allow for the determination of allele or genotype frequency in populations. If a phenotype is determined by a single gene, three genotypes are possible – AA, Aa or aa.

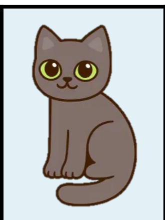
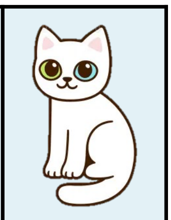
Equation 1: $p + q = 1$

The letter **p** represents the frequency of the dominant allele (A), while **q** represents the frequency of the recessive allele (a). The total frequency of both alleles must always equal 100% for the equation to apply.

Equation 2: $p^2 + 2pq + q^2 = 1$

Because a genotype consists of two alleles, the first equation must be squared to determine the genotype frequencies. This will result in the expanded form of the equation – where p^2 = AA, $2pq$ = Aa and q^2 = aa.

The Hardy-Weinberg model assumes certain conditions must be maintained for a population to remain in genetic equilibrium. The population must be large with **random mating**. There is **no mutation** or gene flow and there is **no natural selection** or allele-specific mortality. If any of these conditions are not being met, the population will not maintain genetic equilibrium and it should be concluded the population is *evolving*.

Phenotypes			
Genotypes	BB	Bb	bb
Genotype Frequency	0.36	0.48	0.16
Allele Frequency	0.36 + 0.24 = 0.6		0.24 + 0.16 = 0.4

Hardy-Weinberg Worked Example:

In a population of 250 cats, 40 cats have white fur (with white fur being recessive to black fur)

How many cats with black fur are heterozygous?

q^2 = **0.16** (40 ÷ 250) q = **0.4** ($\sqrt{0.16}$)

p = **0.6** (1 – 0.4) p^2 = **0.36** (0.6^2)

$2pq$ = **0.48** (2 × 0.6 × 0.4)

Answer: 120 cats (48% of 250)