

Edexcel (B) Biology A-level

Topic 8 - Origins of Genetic Variation

Definitions and Concepts

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8.1 - Origins of genetic variation

Continuous variation - A type of variation within a population produced by polygenic inheritance where the phenotypes are spread over a range of values.

Crossing over - The exchange of genetic material between two chromosomes in a bivalent during meiosis 1.

Discontinuous variation - A type of variation within a population produced by monogenic inheritance which produces phenotypes that can be placed into distinct categories.

Gene mutation - A change to at least one nucleotide base in DNA or the arrangement of bases. Gene mutations occur spontaneously and may result in harmful or beneficial changes to the genotype.

Genetic variation - The difference in the genetic code (DNA) of organisms within a population.

Random fertilisation - The random probability of a specific fertilisation event occurring between two gametes out of a large potential selection of gametes.

8.2 - Transfer of genetic information

Alleles - Different versions of the same gene.

Autosomal linkage - Genes which are usually inherited together as they are on the same non-sex chromosome.

Chi-squared (χ^2) **test -** A statistical test used to determine whether a pattern of inheritance is statistically significant.

Chromosome - A structure composed of tightly condensed DNA which contains many genes.

Codominant - When both alleles for a gene in a heterozygous organism equally contribute to the phenotype.

Dominant trait - A trait which is present if an individual has at least one copy of the allele.

Genetic cross - Combining two organisms to produce offspring with traits from both parents due to the inheritance of genes.

Genotype - The genetic makeup of an organism.

Haemophilia - An X-linked recessive blood disorder which prevents clots from forming properly.

Heterozygote - An organism which has two different versions of the same gene.





Homozygote - An organism which has two of the same versions of a gene.

Linked genes - Genes which have a similar locus and are hence usually inherited together.

Multiple alleles - The presence of more than two genes which can code for a characteristic.

Phenotype - The observable physical characteristics of an organism which is based on both the genotype and environmental influence.

Recessive trait - A trait which is only present when an individual has two copies of the allele and can be masked by a dominant allele.

Sex-linked genes - Genes which are usually inherited together as they are on the same sex chromosome and so expression is sex-dependent.

Unlinked genes - Genes which are at different loci or on different chromosomes which may be subject to independent assortment and are often not inherited together.

8.3 - Gene pools

Directional selection - A type of selection that favours one extreme phenotype and selects against all other phenotypes.

Disruptive selection - A type of selection where multiple extreme phenotypes are advantageous for different reasons and the average phenotype is selected against. This leads to speciation and changes in the population.

Founder effect - A type of genetic drift in which a few individuals of a species break off from the population and form a new colony. This results in smaller gene pools and an increased frequency of rare alleles.

Genetic drift - A gradual change in allele frequencies in a population over time due to chance.

Hardy-Weinberg principle - A principle that states that the frequency of alleles in a population will not change over time unless evolutionary factors are present. It can be used to calculate the frequencies of the other two genotypes when given the frequency of one genotype using the equation given below:

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

 $p^2 =$ Frequency of homozygous dominant

2pq = Frequency of heterozygous

 $q^2 =$ Frequency of homozygous recessive

Population bottlenecks - A significant reduction in population size which reduces the genetic diversity of a population.





Selection pressures - Factors which lead to selection and survival of the fittest, thereby driving evolutionary genetic changes over time.

Stabilising selection - A type of selection which selects against extreme phenotypes and produces a population with average phenotypes.

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