

OCR (B) Biology A-level

5.1.2 - Population genetics and epigenetics

Flashcards

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How does natural selection cause a change in allele frequencies over generations?



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Organisms with advantageous characteristics are more likely to survive and pass their favourable alleles to offspring. Frequency of unfavourable alleles decreases.



Give an example of natural selection affecting allele frequencies in humans.



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- **Carriers** of the sickle cell anaemia gene have some protection against malaria.
- Increases frequency of the heterozygous genotype in malaria-endemic areas.



What effect does a change in amino acid sequence have?



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The amino acid sequences determines the structure and function of the resulting protein. Any change in the sequence will alter the properties of the protein.



What is the Hardy-Weinberg principle?



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Allows us to estimate the frequency of alleles in a population, as well as if allele frequency is changing over time.



Explain the Hardy-Weinberg equation for calculating allele frequency.



Explain the Hardy-Weinberg equation for calculating allele frequency.

The frequencies of each allele for a characteristic must add up to 1.0. The equation is therefore; $p + q = 1$

Where p = frequency of the dominant allele, and q = frequency of the recessive allele.



Explain the Hardy-Weinberg equation for calculating genotype frequency.



Explain the Hardy-Weinberg equation for calculating genotype frequency.

The frequencies of each genotype for a characteristics must add up to 1.0. The equation is therefore;

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

Where p^2 = frequency of homozygous dominant,
 $2pq$ = frequency of heterozygous, and q^2 = frequency of homozygous recessive.



What is meant by a population bottleneck?



What is meant by a population bottleneck?

Where a catastrophic event dramatically reduces the size of a population, thereby decreasing the variety of alleles in the gene pool and causing large changes in allele frequencies.



What is meant by the founder effect?



What is meant by the founder effect?

When a small number of individuals become isolated, forming a new population with a limited gene pool. Allele frequencies not reflective of the original population.



Give examples of the founder effect in the human population.



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- Ellis-van Creveld syndrome more prevalent in Amish communities as they do not interbreed with the general population.
- Blood group distribution varies across different countries.



Define speciation.



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Where a population is split and reproductively isolated, there are different selection pressures on the two groups. If the genetic makeup changes to the extent the two groups can no longer interbreed, they have become separate species.



What types of isolation can lead to speciation?



What types of isolation can lead to speciation?

- Geographical isolation e.g. river, mountain range.
- Reproductive isolation e.g. a mutation that no longer allows two organisms to produce fertile offspring.



What is meant by epigenetics?



What is meant by epigenetics?

A heritable change in gene function
without change to the base sequence of
DNA.



How can histone modification affect gene expression?



How can histone modification affect gene expression?

- Addition of an acetyl group activates chromatin, allowing transcription.
- Addition of a methyl group can either activate or inactivate chromatin depending on the position of the lysine.



How can DNA methylation affect gene expression?



How can DNA methylation affect gene expression?

Involves addition of a methyl (CH_3) group to cytosine bases, which prevents transcription factors from binding. Therefore gene transcription is suppressed.



Discuss research into epigenetics.



Discuss research into epigenetics.

- Dutch Hunger Winter; those affected underwent epigenetic changes which are still passed onto offspring today.
- Twin studies; identical DNA subjected to different epigenetic influences produces variations in phenotype.

