Edexcel Biology GCSE

Topics 3.20 to 3.23 - Variation and the human genome project

Flashcards

What are the two causes of variation within a species?

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- Genetics
- Environment

What is genetic variation?

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- Variations in the genotypes of organisms of the same species due to the presence of different alleles
- Creates differences in phenotypes

What creates genetic variation in a species?

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- Spontaneous mutations
- Sexual reproduction

What is a mutation?

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A random change to the base sequence in DNA which results in genetic variants

State the three types of gene mutation

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- Insertion
- Deletion
- Substitution

How may a gene mutation affect an organism’s phenotype? (3)

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- Neutral mutation does not change the sequence of amino acids. Protein structure and function same. No effect on phenotype.
- Mutation may cause a minor change in an organism’s phenotype e.g. change in eye colour.
- Mutation may completely change the sequence of amino acids. This may result in a non-functional protein. Severe changes to phenotype.

What is environmental variation?

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- Variations in phenotype that are acquired during the lifespan of an organism
- Due to environmental factors e.g. diet, lifestyle, climate, exposure to light etc.

What is the Human Genome Project?

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- Scientific research project involving thousands of scientists across the globe which successfully mapped the entire human genome
- Scientists now aim to identify the function of every gene in the human genome

How can the results of the Human Genome Project be applied to medicine?

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- Enables scientists to understand how lifestyle factors interact with genes - identifying predisposition to disease and possible preventions
- Disease-causing alleles identified more rapidly and the appropriate treatments prescribed earlier on
- Scientists can predict an individual’s response to certain drugs. New drugs can be developed which are tailored to a specific allele.

What are drawbacks associated with the discoveries of the Human Genome Project?

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- Knowledge of predisposition to a disease can be stressful
- Societal pressure influencing the decision to have children
- Discrimination by employers, insurance firms etc.