

## Inheritance - Questions by Topic

Q1. Cystic fibrosis is a genetic disorder caused by mutations in the CFTR gene. One aim of somatic gene therapy is to overcome the effects of defective genes.

(a) (i) Describe the difference between somatic gene therapy and germ line gene therapy.

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\*(ii) Suggest how somatic gene therapy could enable cells lining the lungs to function normally in people with cystic fibrosis.

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(b) Rhythmical tapping of the chest wall during physiotherapy can relieve the symptoms of cystic fibrosis in the lungs.

Suggest an explanation for this.

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**(Total for Question = 8 marks)**

Q2.

Cystic fibrosis is an inherited condition.

Explain why people with cystic fibrosis can have breathing difficulties.

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Q3.

The photograph below shows a cleft iris, a rare condition in humans. Cleft iris may be due to the inheritance of recessive alleles.



Magnification  $\times 1$

Explain the meaning of the term **recessive allele**.

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Q4.

Mutations to DNA can affect the structure of proteins produced in the cell.

Removing one base from a DNA sequence will affect the primary structure of a protein.

Changing one base for another may not affect the primary structure of a protein.

Explain why these two types of mutation have different effects on protein structure.

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Q5.

Leptin is a protein hormone with a role in the control of appetite in humans.

Several mutations of the leptin gene have been identified. All these mutations are frameshift mutations that result in shortened primary structures.

A frameshift mutation involves the insertion or removal of one or two nucleotides from a gene.

Describe how a frameshift mutation could result in the production of leptin with a variety of shorter primary structures.

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