1. Nov/2022/Paper_41/No.1

Adenosine deaminase (ADA) deficiency is an immune system disorder caused by a recessive autosomal mutation.

Severe combined immunodeficiency caused by a lack of ADA is called ADA-SCID.

(a) Genetic engineering is used to make a recombinant human protein to treat people with ADA-SCID.

Outline the principles of genetic engineering.

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(b) In 2016 gene therapy to cure ADA-SCID was approved in Europe. The gene therapy involves three main steps.

- Blood (haematopoietic) stem cells are taken from the bone marrow of the person with ADA-SCID.
- The functional gene and its promoter are inserted into the blood stem cells.
- A single infusion (injection) of the gene-corrected cells is given to the patient.

(i) Explain why a single infusion of gene-corrected stem cells is enough to cure the disease.

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(ii) Explain why a promoter has to be transferred as well as the desired gene.

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(iii) A modified retrovirus is used to insert the new gene into the DNA of the blood stem cells.

State **two** ethical considerations of using a retrovirus for gene therapy.

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(c) The gene therapy technique used to cure ADA-SCID is **not** suitable for treating the genetic disease called Huntington's disease. A newer technique called gene editing could potentially be used instead to cure Huntington's disease.

Explain why gene editing is more suitable as a potential cure for Huntington's disease.

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[Total: 13]

2. June/2022/Paper_42/No.1

(a) In some varieties of domestic cats the gene for fur colour is located on the **X** chromosome. This gene has two alleles. One allele codes for black fur and the other allele codes for ginger fur. The two alleles are codominant so a heterozygous cat will have fur with patches of black and ginger colours. A cat with fur of two colours is known as a tortoiseshell.

Using appropriate symbols, construct a genetic diagram to show the results of a cross between a female tortoiseshell cat and a male ginger cat.

symbols

parent phenotypes tortoiseshell female ginger male

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[5]

(b) In humans the *TYR* gene is involved in the production of a dark pigment, melanin, in some cells.

Describe how the expression of the *TYR* gene leads to the production of melanin.

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[Total: 8]

3. June/2022/Paper_43/No.7

The determination of sex in domestic turkeys is different from that in humans. The sex chromosomes in turkeys are named **Z** and **W**. Male turkeys are **ZZ** and female turkeys are **ZW**.

The gene for feather colour is located only on the **Z** chromosome.

- The dominant allele codes for bronze feathers.
- The recessive allele codes for brown feathers.

(a) Define the terms dominant and recessive.

dominant .....

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recessive .....

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[2]

(b) Using suitable symbols, construct a genetic diagram to show the results of a cross between a heterozygous bronze male turkey and a brown female turkey.

symbols

parent phenotypes bronze male brown female

parent genotypes

gametes

offspring genotypes

offspring phenotypes

[5]