

1 Metachromatic leukodystrophy (MLD) is an inherited disorder. This disorder, due to brain damage during the first two years of life, can prevent a child learning how to walk and talk.

In one of the first gene therapy treatments approved in Europe, one child from each of three families with a history of MLD, was treated.

As a result of this treatment the children were able to talk at an age when some of their untreated brothers and sisters were unable to talk.

MLD is associated with a recessive allele.

(a) Two parents, who are physically unaffected by the disorder, have already had one child with the disorder.

Using a suitable genetic diagram, calculate the probability that the next child of these parents will also be affected by this disorder.

(4)

probability

(b) Suggest how a newborn baby could be tested for MLD before brain damage develops. (2)

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(c) The gene therapy involved taking bone marrow stem cells from the child to be treated. A virus was then used to transfer DNA with the dominant allele into the stem cells. These modified stem cells were then injected into this child.

(i) Suggest how these modified stem cells can result in the prevention of MLD.

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(ii) Suggest why the development of the treated children was compared with the development of their untreated brothers and sisters.

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(d) There are some risks associated with somatic gene therapy.

Suggest why the parents of these children gave consent for their children to be involved in the gene therapy treatment for MLD.

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(Total for Question 1 = 13 marks)

2 In 1886, Sir Francis Galton claimed that the height of a child could be predicted by working out the mean height of its parents.

Scientists have since discovered that the inheritance of height is an example of polygenic inheritance.

(a) Explain what is meant by the term **polygenic inheritance**.

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(b) Some scientists have suggested that diet may affect the growth of a child.

Explain how the height of an adult human demonstrates how environmental factors interact with genotype.

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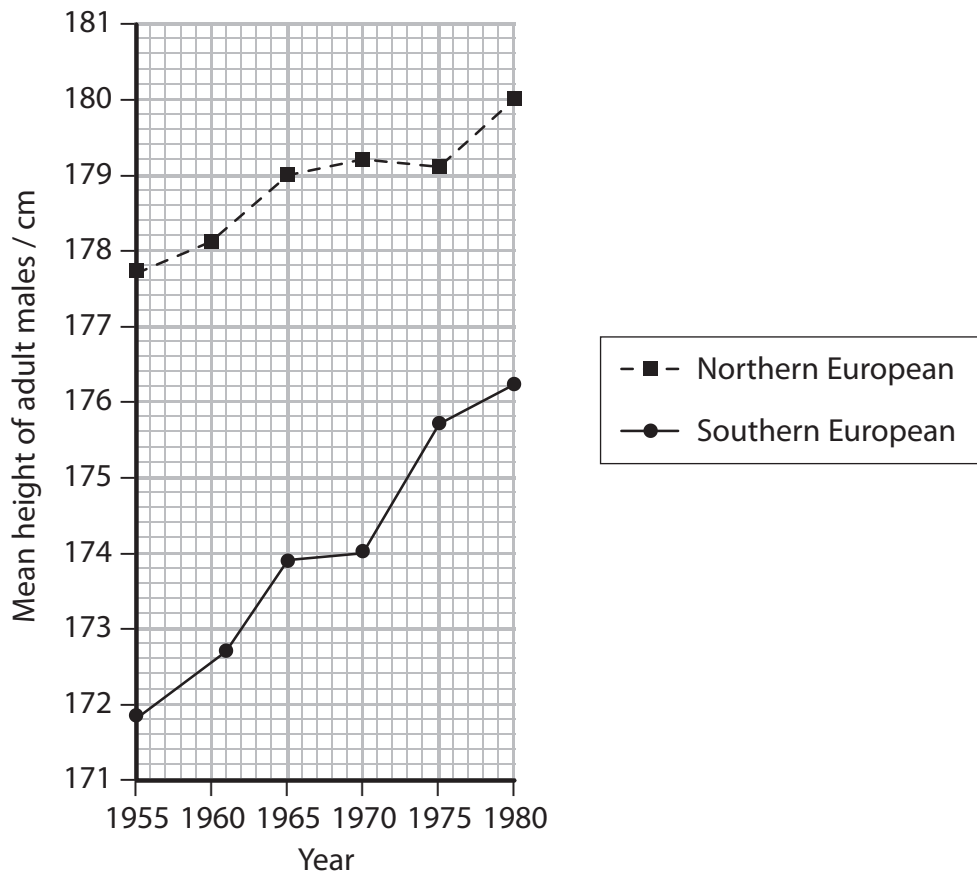
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(c) The graph below shows the mean height of populations of adult males in Northern and Southern Europe from 1955 to 1980.



(i) Using the information in the graph, describe the changes in mean height for adult European males from 1955 to 1980.

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(ii) Suggest an explanation for the changes in mean height of these two groups of adult males.

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(Total for Question 2 = 10 marks)

- 3** The photograph below shows hydrangea flowers. These flowers are white or coloured.

The allele for producing coloured flowers is dominant. If no pigment is produced, the flowers will be white.



Magnification $\times 0.25$

- (a) Suggest the genotype for white hydrangea flowers.

(1)

- (b) The different colours are caused by a pigment. The pigment appears blue when the concentration of aluminium ions in the soil is high. The pigment appears pink when the concentration of aluminium ions in the soil is low.

Scientists investigated the effect of soil pH on the colour of hydrangea flowers.

The table below shows their results.

pH of soil	Flower colour
4.5	Deep vivid blue
5.0	Medium blue
5.5	Lavender purple
6.0	Purplish pink
6.5	Mauve pink
6.8	Medium pink
7.0	Deep vivid pink

- (i) Name the phenotype being investigated.

(1)

- (ii) Using information from the table, explain the effect pH has on the aluminium ions in the soil.

(3)

(iii) Explain why changing the pH of the soil would have no effect on the colour of white hydrangeas.

(2)

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(c) Flowers of other plants show continuous variation in colour due to polygenic inheritance rather than environmental factors.

Explain what is meant by the term **polygenic inheritance**.

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(Total for Question 3 = 9 marks)

4 Scientists have investigated the influence of both nature and nurture on brain development. They used several pairs of identical twins and several pairs of non-identical twins.

(a) In one investigation, each twin was shown a number of human faces and then asked to identify them amongst a group of unfamiliar faces.

The agreement in face identification between each pair of twins was recorded.

The results were used to calculate the mean percentage agreement in face identification for the two types of twin. This is shown in the table below.

Mean percentage agreement in face identification (%)	
identical twins	non-identical twins
70	29

(i) From these results, the scientists concluded that face identification has a genetic component.

Explain how these results support this conclusion.

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- (ii) This investigation was repeated using written words rather than faces. The mean percentage agreement in word identification for the two types of twin suggested that this involved an environmental component.

Suggest how the results of this investigation might differ from the results shown in the table.

(1)

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- (b) Functional magnetic resonance imaging (fMRI) was used in another investigation. Brain activity was recorded whilst carrying out face identification.

Suggest why fMRI was used in this investigation.

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(Total for Question 4 = 9 marks)