1 The central nervous system (CNS) is made up of the brain and the spinal cord.

(a) The image below of a human head and neck shows part of the CNS.

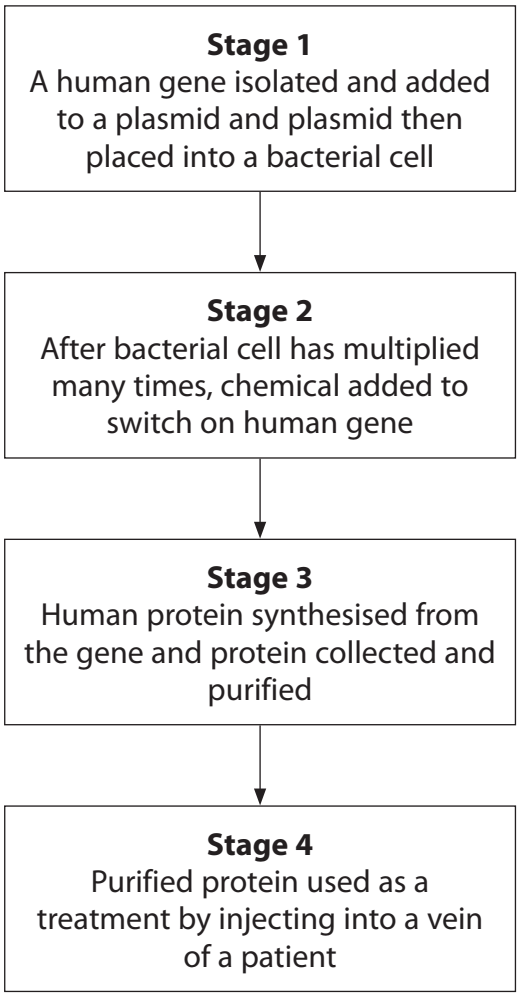
Using the image and your knowledge, complete the table below.

(4)

Labelled structure	Name of structure	One function
A		
		Feel emotions

(b) Some of the drugs used to treat human disorders are proteins. Some of these proteins can be synthesised by genetically modified bacteria.

The diagram below shows some stages in the production of one of these drugs.



(i) A restriction enzyme is used in Stage 1.

Explain why only one restriction enzyme is used in stage 1.

(2)

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(ii) Suggest how the addition of a chemical causes the human gene to be switched on in stage 2.

(3)

(iii) Describe the structure of an organelle found in a bacterial cell that is involved in synthesising human protein in stage 3.

(1)

(iv) Suggest **two** advantages of injecting the protein into a vein rather than an artery in stage 4.

(2)

(Total for Question 1 = 12 marks)

2 Cystic fibrosis and albinism are examples of recessive genetic disorders.

Tay-Sachs disease is another example of a recessive genetic disorder.

(a) Explain the meaning of the term **recessive genetic disorder**.

(2)

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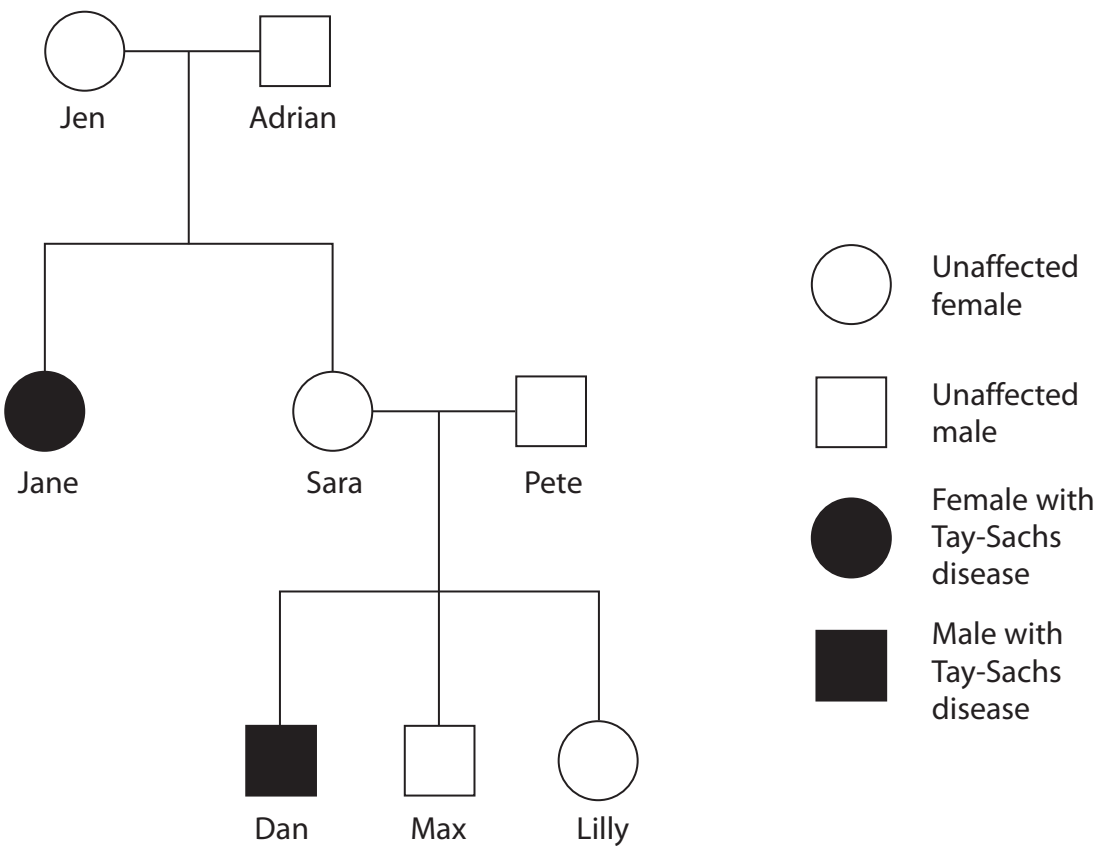
(b) The genetic pedigree diagram below shows the inheritance of Tay-Sachs disease in one family.



For each of the statements below, put a cross (☒) in the box that correctly completes the statement.

(i) The female who definitely has a homozygous genotype is

(1)

☒ **A** Jane

☒ **B** Jen

☒ **C** Lilly

☒ **D** Sara

(ii) The female whose genotype cannot be identified from the diagram is

(1)

☒ **A** Jane

☒ **B** Jen

☒ **C** Lilly

☒ **D** Sara

(iii) A male who definitely has a heterozygous genotype is

(1)

☒ **A** Adrian

☒ **B** Dan

☒ **C** Max

☒ **D** none of them

(iv) A male who definitely is homozygous dominant is

(1)

☒ **A** Adrian

☒ **B** Dan

☒ **C** Max

☒ **D** none of them

*(c) Tay-Sachs disease is caused by a gene mutation that results in the build up of lipid in the brain. It is hoped that gene therapy will be able to treat this disease in the future.

Sheep can also suffer from Tay-Sachs disease. Investigations have found that gene therapy increases the life span of these animals.

Suggest how these gene therapy investigations could have been carried out.

(5)

(Total for Question 2 = 11 marks)

3 The Human Genome Project is helping in the design of new drugs to treat a variety of human diseases and in the development of synthetic tissues.

(a) (i) Explain the meaning of the term **Human Genome**.

(1)

(ii) Describe **one** ethical implication associated with the use of information obtained from the analysis of the human genome.

(1)

(b) Melanoma is an aggressive form of skin cancer.

Very few patients with this cancer survive for more than five years. Some melanomas are associated with a genetic mutation identified by the Human Genome Project.

Drug R (R05185426) has been developed to treat patients with these melanomas. In clinical trials, drug R has been shown to cause a 50% shrinkage of melanomas in only a few months.

(i) Suggest how work on the Human Genome Project helped in the development of drug R.

(3)

(ii) Suggest how drug R may have caused the melanoma to shrink in only a **few months**.

(4)

(iii) Drug R needs one more round of testing, in a phase III trial, before it can be approved for use.

Explain what is meant by a **phase III trial**.

(2)

(c) Yeast cells were genetically modified, using human DNA, to produce collagen. This collagen can be used to make synthetic corneas.

Ten patients who were blind were each given a synthetic cornea. They were all able to see with no reported complications due to tissue rejection.

Suggest why these synthetic corneas were not rejected.

(2)

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(Total for Question 3 = 13 marks)

4 Thalassaemia is the name of a group of inherited blood disorders that affect the body's ability to produce haemoglobin in red blood cells. Red blood cells are produced in bone marrow.

Oxygen in the lungs binds to haemoglobin and is carried to the cells of the body to be used in respiration.

Beta thalassaemia is the result of a mutation in the gene coding for the β chain of haemoglobin. If a person inherits gene mutations from both parents, this person will show symptoms of anaemia and will require blood transfusions. Symptoms of anaemia include tiredness and breathlessness.

*(a) Using the information given above and your knowledge of gene mutation, suggest why a person with beta thalassaemia has symptoms of anaemia.

(4)

(b) If the phenotypes of the parents are known, the probabilities of having a child with beta thalassaemia, an unaffected child or a child who is a carrier, can be calculated.

Complete the table below to show the results of these calculations.

(4)

Parent 1	**Parent 2**	**Probability of having a child with beta thalassaemia**	**Probability of having an unaffected child**	**Probability of having a child who is a carrier**
Unaffected	carrier	no chance	50%	50%
Carrier	carrier			
Unaffected	has beta thalassaemia			
Carrier	has beta thalassaemia	50%	no chance	50%

(c) Gene therapy could potentially be used to treat beta thalassaemia.

Suggest how gene therapy could be carried out to treat this disorder.

(4)

(Total for Question 4 = 12 marks)