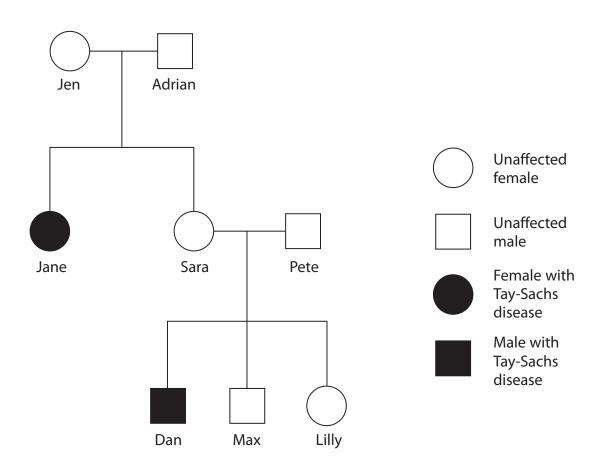
1	Cystic fibrosis and albinism are examples of recessive genetic disorders.	
	Tay-Sachs disease is another example of a recessive genetic disorder.	
	(a) Explain the meaning of the term recessive genetic disorder.	
		(2)

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



		ch of the statements below, put a cross ($oxtimes$) in the box that correctly complete tement.	<u> </u>
(i)	The	e female who definitely has a homozygous genotype is	(1)
X	A	Jane	
X	В	Jen	
X	C	Lilly	
X	D	Sara	
(ii)	The	e female whose genotype cannot be identified from the diagram is	(1)
X	A	Jane	
X	В	Jen	
X	C	Lilly	
X	D	Sara	
(iii)	A n	nale who definitely has a heterozygous genotype is	(1)
X	A	Adrian	
X	В	Dan	
X	C	Max	
X	D	none of them	
(iv)	A n	nale who definitely is homozygous dominant is	(1)
X	A	Adrian	
X	В	Dan	
X	C	Max	
X	D	none of them	

(Total for Question 1 = 11 mar	ks)
	(5)
Suggest how these gene therapy investigations could have been carried out.	(E)
Sheep can also suffer from Tay-Sachs disease. Investigations have found that gene therapy increases the life span of these animals.	
future.	
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	
	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.

Krabbe disease is another example of a recessive genetic disorder. Krabbe disease is caused by mutations in the GALC gene, resulting in a deficiency of an enzyme called galactocerebrosidase.	of
(a) Explain the meaning of each of the following terms.	
(i) Mutation	(2)
(ii) Recessive	(1)
(b) Suggest how a mutation in the GALC gene could result in a change in the enzyr galactocerebrosidase.	me (3)
galactocerebrosidase.	(3)
	(3)
galactocerebrosidase.	(3)
galactocerebrosidase.	(3)

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

(c)	Two parents are both carriers of the recessive allele for Krabbe disease.	
	In the space below, draw a genetic diagram to show the possible genotypes and phenotypes of their children.	
	Use the genetic diagram to find the probability of these parents having a child with Krabbe disease.	(5)
(d)	ProbabilityState how these parents could determine whether or not their unborn child has Krabbe disease.	(1)
	(Total for Question 2 = 12 ma	rks)

3 A study examined the risk of developing a mental disorder. This study determined the risk for both the population as a whole and for those who had a close relative (parent, brother, sister or child) with the same disorder.

The results are shown in the table below.

	Risk of developing mental disorder (%)			
Mental disorder	Population	n as a whole	Those with a close relative	
	Males	Females	with the same disorder	
Alcoholism	7.0	2.0	15	
Anxiety	3.0	6.0	15	
Manic depression	2.0	3.0	15	
Neurotic depression	6.0	12.0	11	
Obsessive compulsive	0.1	0.1	10	
Schizophrenia	1.0	1.0	10	

(a) (i)	People with obsessive compulsive disorder (OCD) have symptoms such as repeated washing, checking, touching, counting or arranging.					
	Using the data in this table, give the evidence that OCD is an inherited condition.	(2)				

(ii) Using the data in the table, explain the validity of the statement that 'OCD is an inherited condition'.		
	an inherited condition.	(2)
(iii)	Using the data in the table, state which disorder is least likely to be an inherited condition.	
	Give a reason for your answer.	(2)
 	/Tatal fan Onastian 3 - Carre	
	(Total for Question 3 = 6 mai	rks)

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)
	(- /

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.

(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%

	(Total for Question 4 = 12 ma	rks)
		(=)
	Suggest how gene therapy could be carried out to treat this disorder.	(4)
(C)	Gene therapy could potentially be used to treat beta thalassaethia.	
(c)	Gene therapy could potentially be used to treat beta thalassaemia.	