1. Some diseases are inherited.

Haemophilia is an example of an inherited disease. It is caused by a **recessive allele**. The gene for haemophilia is located on the sex chromosomes.

Due to the location of the gene for haemophilia, females inherit two copies of the gene, but males only inherit one. For a male, this is shown on the diagram below.



Males cannot be carriers for haemophilia. They either have the disease or they do not.

(i) A female carrier has the genotype X^HX^h and a healthy male has the genotype X^HY.
 Complete the Punnet square to show the probability of this couple having a son with haemophilia.

	X ^H	X ^h
X ^H		
Y		

Probability =_____ [2]

(ii) Which of the following genotypes would a female with haemophilia have?

Put a tick (\checkmark) in the correct box.



[1]

2(a). Scientists use cell structures from three people to make a baby:

- the nucleus from a mother's egg cell
- the nucleus from a father's sperm cell
- the mitochondria from a donor's egg cell.

This technique will help prevent some genetic diseases caused by faulty mitochondria.

The diagram below shows how the process will be done.



Step 4 Fertilised egg cell is then placed in the mother's uterus.

Mitochondria contain 37 genes.

The nucleus of a fertilised egg cell contains 40 000 genes.

What percentage of its genes does the fertilised egg cell receive from the donor?

Give your answer to 2 decimal places.

Show your working.

_____ % [2]

(b). Most of the baby's physical characteristics will be inherited from its father and mother.

Suggest why.

______[1]

(c). Genes code for proteins.

What type of protein could the genes in the mitochondria code for?

.....[1]

(d). Babies created by this new technique will contain the DNA from 3 different individuals.

Some people do not agree with the use of this new technique.

Suggest and explain why.

[3]

(e). Approximately 1 in 200 children have faulty mitochondria.

1 in 6500 children will have serious diseases as a result.

Do you think this justifies the development of this new technique?

Explain your answer.

[2]

(f). The DNA in the mitochondria of people affected by mitochondrial disease contains mutations.

A mutation is a change in the base sequence of the DNA.

Explain how these mutations can cause problems.

[2]

3. Jack and Ted are identical twins.

Identical twins are an example of naturally occurring clones.



How many egg cells and sperm cells were needed during fertilisation to produce these identical twins?

Put a tick (\checkmark) in the box next to the correct answer.

Two egg cells and two sperm cells.	
Two egg cells and one sperm cell.	
One egg cell and two sperm cells.	
One egg cell and one sperm cell.	

4(a). Our genes and chromosomes contain genetic information.

Write a word in the gap to complete each sentence.

Genes are instructions for a cell that describe how to make _____.

Genes are sections of long molecules of _____ that make up chromosomes.

The combination of alleles an organism has is called its _____.

The observable characteristics of an organism are called its _____.

(b). Human body cells usually contain 23 pairs of chromosomes.

In males and females, 22 of these pairs of chromosomes look the same.

Write down the name of the pair of chromosomes that look different in males and females.

----·

[2]

5. Cathy is pregnant with triplets.

All three babies have the same father.

When the babies are born, two of the babies, A and B, are genetically identical to each other.

The third baby, C, is genetically different.

Explain why babies A and B are genetically identical and why baby C is not genetically identical to babies A and B.

The quality of written communication will be assessed in your answer.

[6]

6. Harold has cystic fibrosis.

Hilda is a carrier for the disease.

(i) Write down Harold and Hilda's genotypes.

Use F to represent the dominant allele and f to represent the recessive allele.

Harold's genotype:	Hilda's genotype:	[1]

(ii) Which row in the table correctly describes Harold and Hilda's genotypes?

Put a tick (\checkmark) in the box at the end of the correct row.

Harold	Hilda		
heterozygous	homozygous dominant		
heterozygous	homozygous recessive		
homozygous dominant	heterozygous		
homozygous dominant	homozygous recessive		
homozygous recessive	homozygous dominant		
homozygous recessive	heterozygous		

7. Explain how a sample of fetal cells can be used to find out the sex of a fetus.

[2]

[1]

8. Harold has cystic fibrosis.

Hilda is a carrier for the disease.

Hilda is pregnant.

Harold is the father of the unborn baby.

Harold thinks that, because he has cystic fibrosis, the baby will definitely have cystic fibrosis.

Explain why Harold is not correct.

[2]

9. Explain how a gene can determine the sex of a fetus.

 [3]

10. Cystic fibrosis is an inherited disorder.

Sharon and Eric are both carriers for cystic fibrosis.

Sharon is pregnant. Eric is the father.

There is a chance that the fetus will have cystic fibrosis.

Calculate the probability that the fetus will have cystic fibrosis.

Use a genetic diagram or Punnett square to help you.

Use T = normal allele and t = cystic fibrosis allele.

probability of fetus having cystic fibrosis = _____ [3]

11(a) Humans usually have two alleles for each gene.

Genes determine the characteristics shown by the person.

Draw a straight line to connect each word with its correct definition.



[3]

- (b). Marfan syndrome is a genetically inherited disorder.
 - (i) Look at the genetic diagram.



Marfan syndrome is caused by a dominant allele. Complete the diagram to show the inheritance of Marfan syndrome.

Use G for the dominant allele and g for the recessive allele.

(ii) Write down the three possible combinations of genotypes for the parents of individual 6.

Combination 1 _____ and _____

Combination 2 _____ and _____

Combination 3 _____ and _____

[2]

[3]

(c). Janet and John are expecting a baby.

John has mild Marfan syndrome.

Marfan syndrome can produce a range of conditions from mild (long limbs and fingers) to severe (heart and lung defects).

Janet and John need to decide whether to have the fetus tested for Marfan syndrome.

Write down **three** things that Janet and John need to consider before deciding whether to have the test for Marfan syndrome.

[3]

12. Ali and Mary do not have cystic fibrosis, but their baby does.

What does this tell us about Ali and Mary's genes for this disorder?

]]	<u>1]</u>

13. Plants can be infected by diseases caused by pathogens.

The plant disease ash dieback was first recorded in the early 1990s in Poland.

Since then, many thousands of trees in northern Europe have become infected.



Ash dieback was first found in eastern parts of Great Britain (GB) in 2012, and has been spreading across the country ever since.

The British outbreak of ash dieback started in woodland in Norfolk.

Much of the woodland has died, but one ash tree has shown tolerance to the disease. This tree was named 'Betty' by scientists. Tolerant trees have also been found in mainland Europe.

(i) Explain how scientists could use selective breeding to produce ash trees with improved tolerance.

	[3]
(ii)	New woodland could be planted using cuttings from Betty.
	Explain why this could be a disadvantage during a future outbreak of plant disease.
	[2]
(iii)	Explain how gene technology could be used to produce ash trees with improved tolerance.

14. The Galapagos Islands are a group of 13 islands found in the Pacific Ocean.

Charles Darwin visited the Galapagos Islands during the 19th century.

He collected samples and made observations.

This work helped Darwin to develop a new explanation for the evolution of species.

(i) Which of the following are observations made by Darwin?

Tick (\checkmark) two boxes.

There are differences between fossils and living examples of similar organisms.

Pea plants with red flowers can produce offspring with white flowers.

There is usually extensive variation within a population of a species.

Some bacteria have become resistant to antibiotics.

Isolated populations of the same species living in different places have different characteristics.

[2]

[1]

(ii) Darwin suggested a theory to explain his observations.

Write down the name of the theory he suggested.

15(a) A gene affects whether people have dimples in their cheeks. There are different variants of this gene.

An individual with the dominant variant, D, of this gene will have dimples.

Jack and his wife Nina both have dimples.

Their daughter Mia does not have dimples.

(i) Complete the table to show the genotype of each individual.

Individual	Genotype
Jack	
Nina	
Mia	

[3]

(ii) Jack and Nina decide to have another child.

What is the probability that the second child will have dimples?

Use the Punnett square to show your working.

(b). Scientists consider this trait an 'irregular' dominant trait. This is because sometimes a person can have dimples but their children do not.

What could be responsible for this difference?

.....[1]

END OF QUESTION PAPER

Question		n	Answer/Indicative content	Marks	Guidance
1		i	Correct Punnet square \checkmark X^HX^HX^hX^HX^HX^HX^HX^hYX^HYX^hYProbability 25% / ¼ / 1 in 4 \checkmark	2	ECF correct probability if Punnet square
		ii	X ^h X ^h ✓	1	
			Total	3	
2	a		40,037 / 40,000 + 37; 0.09;	2	0.09 must be expressed to two decimal places. Examiner's Comments Given it was a calculation, this was answered relatively poorly with the majority of candidates gaining no marks. Of those who scored, many gave 0.09 as the answer, but with no working or the incorrect working. Many candidates got the answer 0.09 but often by carrying out the calculation (37/40000) × 100, which limited them to one mark. It is important to note that showing working here was essential to gain the second mark, and candidates should always be encouraged to show their working. Some candidates gave the answer to more than two decimal places, or to two significant figures. Many candidates calculated 40000/37.

Question	Answer/Indicative content	Marks	Guidance
b	 any one from: majority of / most of / 99.91% of their genes/chromosomes/genetic information/DNA from the mother and father/parents/sperm and egg/fertilised egg (not the donor); only small percentage of their genes/chromosomes/genetic information/DNA inherited from the donor; idea that most characteristics are coded for by DNA/genes/chromosomes/genetic material found in the nucleus; 	1	ignore reference to 50% from mother / 50% from father Examiner's Comments This question was answered poorly, with candidates not taking time to understand what was being asked. Many candidates focused on half of genes/23 chromosomes originating from each parent, without credit. Although some candidates had the correct idea that most genes come from the mother and father, they frequently forgot to say 'most', failing to understand what the question was actually asking. Some did say that characteristics are coded for by genes found in the nucleus. Very few made explicit the idea that few genes originated from the donor.
c	enzymes	1	 ignore named proteins / enzymes accept structural/structure / functional/function (proteins) Examiner's Comments Most candidates did not score on this question, with a surprising number of no response answers. Of those who did score, enzymes was frequently given, with some candidates giving functional as a response and very rarely structural. Some did name specific enzymes or proteins, such as e.g. amylase/keratin and some candidates wrote 'hair', but these were not worthy of credit. Many also wrote 'amino acids', but again this was not worthy of credit. Given enzymes are a type of protein, the proportion of wrong answers was surprising.

Question	Answer/Indicative content	Marks	Guidance
e	any two from (1 in 200 is a) high number of children affected; (so) less money will be spent treating children with diseases; (so) prevents faulty mitochondria being passed on to offspring/children; (but) low number (seriously) affected / only 1 in 6,500 / small chance of being (seriously) affected (so it may be) cheaper to treat those affected (than to develop the new technique); (however) idea that money used for the treatment only benefits few people / one disease / could benefit more patients/other diseases; it is worth it even to save one life / improve the quality of life / health;	2	ignore reference to religious and ethical arguments accept alternative idea that this is a high number in a whole population Examiner's Comments This question frequently scored 1 out of the 2 marks, with a significant number of candidates failing to use the information provided in the question. Many candidates gained credit for stating that the technique would improve quality of life, or save lives. Many candidates identified 1 in 6500 being a low number. Very few candidates discussed the idea of preventing faulty mitochondria being passed on. Some candidates did use both the 1 in 200 and the 1 in 6,500 figures thoughtfully in their answers to score 2 marks. Few candidates referred to it being cheaper to treat those affected than to develop the new technique.

Question		n	Answer/Indicative content	Marks	Guidance
Qu	f	n	Answer/Indicative content any two from amino acid sequence will be different/ the amino acids coded for will be different; no/different/incorrect protein/enzyme produced; protein/enzyme may not function;	Marks 2	Guidance ignore changes to the production/formation of amino acids Examiner's Comments Generally the idea that a different or wrong protein would be produced was scored by many candidates. The concept of amino acid sequence being changed seemed not to be so well understood, and rarely scored. There were quite a few references
					to amino acid <i>production</i> , which on its own did not gain credit. Quite a lot of candidates seized on the mutation idea and described how a mutation could affect an individual ranging from various disabilities to cancer, occasionally also talking about incorrect base pairing. The link between a protein being different and a protein not functioning was not often seen, so relatively few candidates scored the final marking point.
			Total	11	
3			One egg cell and one sperm cell. ✓	1	two or more ticks = 0 marks Examiner's Comments This question was well answered. The majority of candidates knew the process of fertilisation to produce identical twins.
			Total	1	

Question		n	Answer/Indicative content	Marks	Guidance
4	а		proteins:	2	four correct = 2 marks two or three correct = 1 mark one correct = 0 marks accept enzymes
			DNA; genotype; phenotype		Examiner's Comments Many candidates were able to provide at least 2 correct responses for 1 mark.
	b		sex (chromosomes) / XX / XY / X AND Y	1	do not credit 'sex cells / gametes' / genes do not credit 'X' alone or 'Y' alone Examiner's Comments The majority of candidates were able to produce the correct response.
			Total	3	

Question	Answer/Indicative content	Marks	Guidance
5	[Level 3] Explanation of how A & B are produced AND explains why C is different to the other two using some level 3 responses. Quality of written communication does not impede communication of the science at this level.	6	This question is targeted at grades up to A* Indicative scientific points may include: A and B Level 1/2 responses
	[Level 2] Explanation of how A & B are produced AND recognises that C must be from a different sperm / egg (combination). Quality of written communication partly impedes communication of the science at this level.		 from the same fertilised egg embryo / zygote / fertilised egg splits / cells separate same genes / genetic information Level 3 responses
	(3 – 4 marks) [Level 1] Explanation of how A & B are produced OR recognises that C must be from different sperm / egg (combination). Quality of written communication impedes communication of the science at this level. (1 – 2 marks) [Level 0] Insufficient or irrelevant science. Answer not worthy of credit. (0 marks)		 identical DNA / chromosomes / alleles / genotype cell division / mitosis creates identical twins / natural clones C Level 1/2 responses formed from different sperm / egg / embryo genes from both parents come together Level 3 responses
			 variation in offspring inherit different combinations of alleles some genes / DNA / chromosomes will be the same / some different ignore reference to egg or sperm splitting Examiner's Comments Many excellent responses contained detailed descriptions on the origins of the identical twins A and B and the non- identical twin C. Others needed to discuss the differences between C and the identical twins in terms of alleles to gain the higher

Question		n	Answer/Indicative content	Marks	Guidance
					marks.
			Total	6	
6		i	Harold: ff Hilda: Ff	1	 need both for the mark allow fF for Hilda do not credit if the distinction between the capital letter and the small letter is not clear Examiner's Comments Many candidates were able to provide the two correct genotypes. The distinction between the capital letter and the small letter had to be unambiguous to gain the mark. no ecf from ai
			heterozygous homozygous dominant heterozygous homozygous recessive homozygous dominant heterozygous homozygous dominant heterozygous homozygous dominant homozygous recessive homozygous recessive homozygous dominant homozygous recessive homozygous dominant homozygous recessive homozygous dominant homozygous recessive homozygous dominant		If more than 1 box is ticked, no mark awarded <u>Examiner's Comments</u> The majority of candidates were able to identify the correct response.
			Total	2	
7			(look at the sex) chromosomes / Karyotype (1) males: XY and females: XX (1)	2	Males have XY chromosomes and females XX chromosomes = 2 marks Examiner's Comments Good responses linked chromosomes to the correct gender. Answers using genes or DNA did not get the mark.
			Total	2	

Question		n	Answer/Indicative content	Marks	Guidance
8			Any two from idea of F / dominant allele being inherited from Hilda / Harold will only pass on one f allele (1) so child might be heterozygous / carrier / Ff (1) there is a 50% chance (1) need 2 recessive alleles to have cystic fibrosis / homozygous recessive / ff (1)	2	Ignore Hilda is a carrier Points could be awarded from a correctly annotated diagram. Do not credit gene instead of allele Examiner's Comments Good responses demonstrated clear understanding of how cystic fibrosis is inherited and were able to prove why the statement was incorrect.
			Total	2	
9			gene on Y chromosome / SRY gene (1) leads to formation of testes / testosterone / androgen (1) absent leads to formation of ovaries / female reproductive system (1)	3	'hormone' alone is insufficient Examiner's Comments This was a challenging question. Candidates needed to know about the sexdetermining gene on the Y chromosome and how it has its effect on gender.
			Total	3	

Question		Answer/Indicative content				Marks	Guidance	
10			T t Probabili	T TT Tt ty = 0.25	t Tt tt		3	one mark for correct parent genotypes (both Tt / tT) incorrect genotypes do not credit for ecf marks one mark for correct completion of Punnett Square. Allow correct alternative genetic diagram. one mark for correct probability Use of alternative symbols (as long as upper and lower case of same letter is clear). Max 2 marks. allow 1 / 4 or 25% or 1 in 4 or 1:3 ignore 1 in 3 Examiner's Comments This was a well answered question. Many candidates were able to produce a correct genetic diagram and probability. Some candidates limited their mark by not using the letters provided.
			Total				3	

Q	uestio	n	Answer/Indicative content	Marks	Guidance
11	а		dominanttwo alleles of a gene that are differentgenotypethe genetic makeup of an organismheterozygousan allele that always shows an effect in the organismhomozygousan allele that only shows an effect if both alleles of the pair are the samephenotypethe observable characteristics of an organismrecessivetwo alleles of a gene that are the same	3	all correct = 3 marks 4 or 5 correct = 2 marks 3 correct = 1 mark Examiner's Comments The majority of candidates scored well on this question, when marks were lost is was generally because of confusion between the definitions of dominant and recessive
	b	i	gg Gg; gg gg Gg gg; Gg gg;	3	 1 mark for each correct row if no fully correct rows allow 1 mark for all homozygous recessives (gg) correct accept alternative letters if clearly upper and lower case used correctly Examiner's Comments This proved to be a difficult question, with only the strongest candidates gaining all three marks; as similar questions where punnet squares have to be completed generally score well perhaps candidates have less experience of unpicking family trees. A large number of candidates only wrote one allele in the spaces provided, others correctly identified the double recessive individuals but then lost marks for not realising all the individuals with Marfan syndrome were heterozygous.

Question		n	Answer/Indicative content	Marks	Guidance
		ii	gg X gg Gg X Gg Gg X gg OR gg X Gg	2	combinations can be in any order accept Gg either way round (Gg or gG) all correct = 2 marks 2 correct = 1 mark Examiner's Comments The majority of candidates wrote the parental genotypes as one allele/letter only so gained no marks. Even those candidates who correctly wrote genotypes often lost a mark for not realising that "gg and Gg" is the same as "Gg and gg". Some wrote numbers from the genetic diagram suggesting they had misread the question.
	C		risk of test eg risk of miscarriage / risk of infection / harm to fetus / risk to mother; will they terminate / abort; false negatives and positive results / test is not 100% accurate;	3	must have the idea of harm to mother or fetus do not allow "affect" fetus accept idea of not keeping the fetus do not credit references to ethical or financial considerations Examiner's Comments Many candidates were aware that the test carried a risk and the result would require a decision about termination, although answers here were often too vague to score e.g. "what will they do with the baby". Fewer candidates recognised that the tests could produce false results. A significant number of candidates discussed ethics, job prospects, insurance premiums which were not appropriate answers to this specific question.
			Total	11	

Question		n	Answer/Indicative content	Marks	Guidance
12			(both Ali and Mary) they are carriers / it is recessive / have faulty gene or allele;	1	accept heterozygous reject Ali OR Mary Examiner's Comments Most candidates scored this mark for correctly stating that both Ali and Mary were carriers. The most common errors were stating that either Ali or Mary was a carrier or providing an incorrect genotype.
			Total	1	

Qı	Question		Answer/Indicative content	Marks	Guidance
13		i	cross Betty with other tolerant/resistance trees / breed two tolerant or resistant trees ✓ test/identify the offspring for tolerance/resistance to ash dieback ✓ select/breed the (most) tolerant/resistance offspring (and repeat) ✓	3 (AO 2.1 × 3)	Examiner's Comments In the slightly unfamiliar scenario of plants rather than the animals, candidates did not explain the process of selective breeding and gave responses involving taking cuttings, cloning or genetic engineering. The key words 'selective breeding' appear to have been ignored.
		ан алан алан алан алан алан алан алан а	the cuttings will be genetically identical to Betty / will have no genetic variation ✓ could all have (a variant/allele that codes for) susceptibility to a different disease/pathogen ✓	2 (AO 2.1) (AO 1.1)	ALLOW will be clones of Betty but DO NOT ALLOW will be similar to Betty /have the same genes as Betty Examiner's Comments Many candidates were able to recognise the fact that clones were genetically identical and a number then went on to explain that they would be susceptible to another disease (rather than the same one). Additionally there was some confusion between resistance of the tree and of the pathogen. A common misconception was the use of the term immunity in relation to the tree.

Question		n	Answer/Indicative content	Marks	Guidance
		iii	sequence the genomes of Betty / other tolerant trees ✓ look for variants/alleles/sequences they have in common ✓ isolate/replicate variants/alleles/sequences associated with tolerance/resistance ✓ use genetic engineering to introduce	4 (AO 2.1 × 4)	ALLOW use of (restriction) enzymes for this process ALLOW description of a method of genetic
			tolerance/resistance (variants/alleles/sequences) into new ash trees ✓		engineering Examiner's Comments This longer response question was designed to assess candidates' knowledge of the whole process of genetic engineering, set in a slightly different scenario. Higher ability candidates produced responses which included sequencing of genomes and the use of genetic engineering.
			Total	9	
14		i	There are differences between fossils and living examples of similar organisms√ Isolated populations of the same species living in different places have different characteristics √	2 (AO 1.1 x 2)	
		ii	natural selection ✓	2 (AO 1.1)	ALLOW survival of the fittest Examiner's Comments Question (a) (i) and (a) (ii) tested candidate knowledge of the work carried out by Darwin in the development of his theory of evolution by natural selection. A common error in (a) (i) was the selection of the statement 'There is usually extensive variation within a population of a species'. In (a) (ii) candidates often only used the stem of the question to state the theory was evolution, rather than natural selection.
			Total	3	

Question			Answer/Indicative content				Marks	Guidance
15	а	i	Jack genotype Dd ✓ Nina genotype Dd ✓ Mia genotype dd ✓				3 (AO 2.1 x 3)	ALLOW other letters in place of D, allow correct descriptions of genotype – heterozygous and homozygous recessive
	ii						2 (AO	ALLOW ECF
				D	d		2.1)	All four genotypes must be correct for first mark
			D	DD	Dd			
			d	Dd	dd			
			Probability = 0.75 ✓				(AO 3.2b)	ALLOW 3 in 4, ³ / ₄ , 75%
								Examiner's Comments
								On the whole this question was answered very well. With a large proportion of candidates gaining 5 or 6 marks for the entire question. The most common errors observed in 7(a) (ii) saw candidates stating the probability as 25% rather than 75%.
	b		Any one from:				1 (AO	Evaminar's Comments
			both parents are heterozygotes ✓ other genes/ mutations ✓				2.1)	
								On the whole this question was answered very well. With a large proportion of candidates gaining 5 or 6 marks for the entire question. 7 (b) was the question that proved more difficult, a common mistake was to state a parent, rather than both parents would be heterozygous
			Total				6	