

1. In humans, sex is determined by chromosomes.

Write down the combination of sex chromosomes in the body cells of females and males.

Females -----

Males -----

[1]

2.

- (i) Some human diseases are not caused by microorganisms, they are inherited. Cystic fibrosis is an example of a disease that is inherited. It is caused by a recessive allele.

Cystic Fibrosis alleles F = dominant f = recessive

Which of the following genotypes would result in the person being affected by cystic fibrosis?

Put a tick (✓) in the correct box.

| | |
|----|--------------------------|
| FF | <input type="checkbox"/> |
| Ff | <input type="checkbox"/> |
| fF | <input type="checkbox"/> |
| ff | <input type="checkbox"/> |

[1]

- (ii) Two parents have a genotype Ff.

Work out the probability of them having a child with cystic fibrosis.

| | | Mother | |
|--------|---|--------|---|
| | | F | f |
| Father | F | | |
| | f | | |

Probability _____

[2]

3(a). Albinism is an inherited condition where affected people are unable to make a pigment called melanin. Skin, hair and eyes may all be affected and the person will be very pale skinned with white-blonde hair and possibly red eyes.

Esther and Simon's daughter, Livvy, has albinism.

Esther and Simon are both heterozygous (carriers).

Complete the Punnett square below to show how Esther and Simon passed the alleles for albinism to Livvy.

Use **A** to represent the allele for normal melanin production and **a** to represent the allele for albinism.

Livvy's genotype is **aa**.

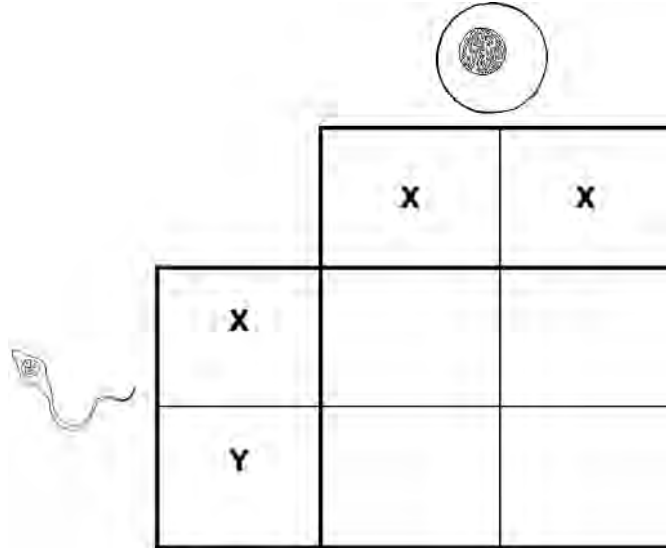
| | | |
|--|----------|----------|
| | A | a |
| | | |
| | | |

[2]

(b). In humans, sex chromosomes determine gender.

Esther and Simon are having another child.

Use the diagram below to show the probability of Esther and Simon's second child being a boy.



Probability -----

[2]

(c). Use the example of the inheritance of albinism to describe the difference between **homozygous** and **heterozygous**.

[2]

4(a). Jane goes to her doctor to have a genetic test.



The doctor finds that Jane has a faulty allele.

Women with this faulty allele are at greater risk of cancer.

The doctor tells Jane there is an 87% chance she will develop breast cancer.

(i) What is the **probability** that Jane will develop breast cancer?

Draw a **ring** around the correct answer.

1 in 87

0.13

0.87

87

[1]

(ii) What would it mean for Jane if her probability of developing breast cancer was 1?

----- [1]

(b). Jane could have major surgery to reduce her risk of developing breast cancer.

The surgery would remove tissue from Jane's body.

What must she consider when deciding whether or not to have the surgery?

----- [3]

(c). Jane can never be free from the risk of cancer.

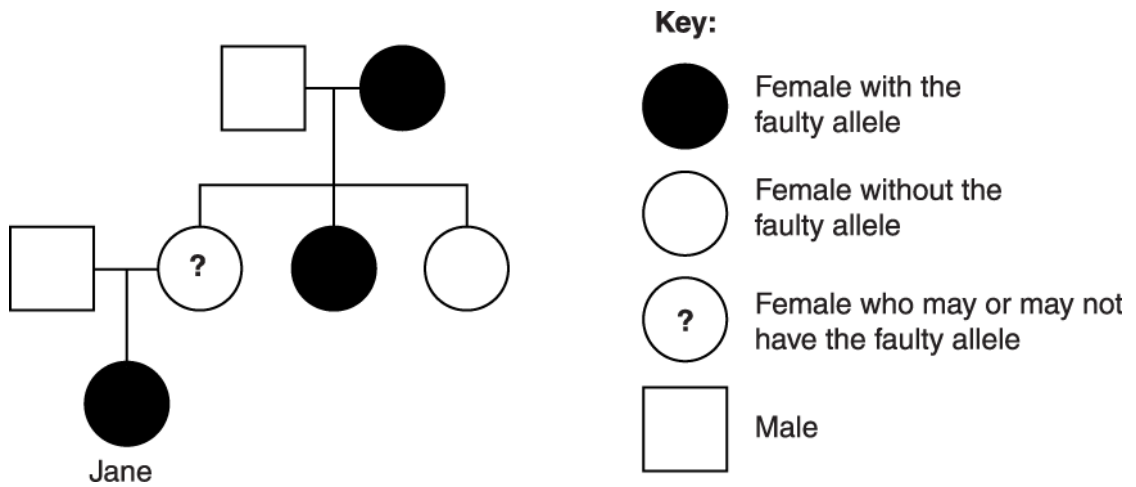
Suggest why.

----- [1]

(d). Jane's doctor looks at information about the allele and at part of Jane's family tree.

Information about the allele:

The normal allele can become faulty during a person's life. This happens in one person out of every 1000.



The doctor concludes that Jane's mother probably has the faulty allele.

What evidence supports this conclusion?

Your answer should include evidence from the family tree and from the information about the allele.

----- [2]

5. Which cells in a human usually contain **pairs** of chromosomes?

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

all human cells

human body cells

human egg cells

human sperm cells

[1]

6. 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?

----- [1]

7(a). Steve and Jane are expecting a baby.

Jane's sex chromosomes are XX.

Complete the Punnet square to show the possible combinations of sex chromosomes in their baby.

| | | Jane | |
|-------|--|------|---|
| | | X | X |
| Steve | | | |
| | | | |

[2]

(b). Steve and Jane already have two baby boys.

Steve says there is a higher chance that the new baby will be a girl.

Explain why Steve is wrong.

[2]

9. Every person has two alleles for a gene.

These alleles can be dominant or recessive.

Draw one straight line from each pair of alleles to the characteristic that the person would have.

Pair of alleles

two dominant alleles

one dominant and one recessive allele

two recessive alleles

Characteristic

the dominant characteristic

the recessive characteristic

[2]

10(a) Write down the combination of sex chromosomes in the body cells of human males and females.

males _____

females _____

[1]

(b).

(i) In the UK, the expected ratio of male to female births is 1 : 1.

2000 babies are born at one hospital in a year.

How many of these would you expect to be female?

answer = _____ [1]

(ii) In another country, the ratio of males to females born is 1.2 : 1.

1000 females are born in a day.

Calculate how many males you would expect to be born on the same day.

Show your working.

answer = _____ [2]

(iii) Suggest why the ratio of males to females born in some countries is 1.2 : 1.

----- [2]

11(a) Cystic fibrosis is an inherited disorder.

Sharon and Eric are both carriers for cystic fibrosis.

Sharon is pregnant. Eric is the father of the baby.

(i) Complete the diagram to show the possible combinations of alleles for their baby.

Key

T = normal allele

t = cystic fibrosis allele

| | | Sharon | |
|------|-------|--------|-------|
| | | T | |
| Eric | T | | |
| | | | |

[2]

(ii) Calculate the probability that the baby will have cystic fibrosis.

probability of baby having cystic fibrosis = [1]

(b). Sharon and Eric discuss whether to have their fetus (unborn baby) tested.

This is what they discuss.

- A The results of the test might not be accurate.
- B The test will enable us to plan treatment if the fetus has cystic fibrosis.
- C Cystic fibrosis is a serious disease.
- D We would rather not know whether our baby has cystic fibrosis.
- E The test is painful for the mother.

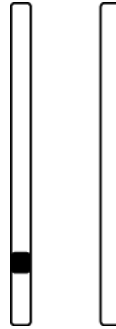
Sharon and Eric decide to have their fetus tested.

Use the ideas of benefit and risk to suggest why they made that decision.

[2]

13. The gene that controls polydactyly is shown on one of the chromosomes below.

(i) Draw the gene in the correct position on the second chromosome of the pair.



[1]

(ii) What is the maximum number of alleles one person can have for this gene?

Put a ring around the correct answer.

0

1

2

23

46

[1]

14. A difference between humans is whether they are male or female.

A student looks at a Punnett square that shows inheritance of sex.

| | | female | |
|------|---|--------|----|
| | | X | X |
| male | X | XX | XX |
| | Y | XY | XY |

The student makes these two conclusions.

- | |
|--|
| <ol style="list-style-type: none">1. Out of 100 babies born, 50 will be male and 50 will be female.2. If a mum and dad have two girls there is a 25% chance that the next baby will be a boy. |
|--|

State whether the conclusions are correct and explain your answer.

[3]

15. Jack dislikes the taste of sprouts. He thinks they taste bitter. His partner Nina loves the taste of sprouts.

Jack reads that a gene affects how people taste sprouts. There are several variants of this gene.

An individual with the dominant variant, T, can taste a bitter substance in sprouts.

(i) Jack is homozygous for this gene.

What is Jack's genotype?

Tick (✓) **one** box.

TT

Tt

tT

tt

[1]

(ii) Jack wants to know if any of his children will be able to taste the bitter substance.

Nina has the genotype tt.

Complete the Punnett square to show the possible genotypes of any children Jack may have with Nina.

| | | |
|--|--|--|
| | | |
| | | |
| | | |

[2]

(iii) What is the probability that any children born will be able to taste the bitter substance.

Probability = [1]

(iv) Jack and Nina have two children, one boy and one girl.

Describe how sex is determined in humans.

.....
.....
.....
..... [2]

(v) Jack and Nina do not want any more children.

They have considered different forms of contraception.

Suggest a form of contraception that would be suitable for them and justify your choice.

.....
.....
.....
..... [2]

END OF QUESTION PAPER

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance | | | | | | | | | |
|----------|-----------|-----------|---|----------|--|----------|----------|-----------|-----------|----------|-----------|-----------|---|---|
| 1 | | | Female XX, male XY ✓ | 1 | Both answers need to be correct for one mark | | | | | | | | | |
| | | | Total | 1 | | | | | | | | | | |
| 2 | | i | ff ✓ | 1 | If more than one box is ticked, do not award the mark even if the correct box is also ticked | | | | | | | | | |
| | | ii | Punnett square correct ✓ <table border="1" style="margin-left: auto; margin-right: auto;"> <tr> <td></td> <td>F</td> <td>f</td> </tr> <tr> <td>F</td> <td>FF</td> <td>Ff</td> </tr> <tr> <td>f</td> <td>Ff</td> <td>ff</td> </tr> </table> ¼ / 0.25 / 25% ✓ | | F | f | F | FF | Ff | f | Ff | ff | 2 | ALLOW ff if given instead of Ff |
| | F | f | | | | | | | | | | | | |
| F | FF | Ff | | | | | | | | | | | | |
| f | Ff | ff | | | | | | | | | | | | |
| | | | Total | 3 | | | | | | | | | | |
| 3 | a | | <table border="1" style="margin-left: auto; margin-right: auto;"> <tr> <td></td> <td>A</td> <td>a</td> </tr> <tr> <td>A</td> <td>AA</td> <td>Aa</td> </tr> <tr> <td>a</td> <td>Aa</td> <td>aa</td> </tr> </table> | | A | a | A | AA | Aa | a | Aa | aa | 2 | One mark for correct gametes for second parent One mark for correct completion of Punnett square ALLOW aA for Aa |
| | A | a | | | | | | | | | | | | |
| A | AA | Aa | | | | | | | | | | | | |
| a | Aa | aa | | | | | | | | | | | | |
| | b | | Punnett square correct ✓ <table border="1" style="margin-left: auto; margin-right: auto;"> <tr> <td></td> <td>X</td> <td>X</td> </tr> <tr> <td>X</td> <td>XX</td> <td>XX</td> </tr> <tr> <td>Y</td> <td>XY</td> <td>XY</td> </tr> </table> Probability is 50% / 0.5 / ½ ✓ | | X | X | X | XX | XX | Y | XY | XY | 2 | ALLOW YX for XY |
| | X | X | | | | | | | | | | | | |
| X | XX | XX | | | | | | | | | | | | |
| Y | XY | XY | | | | | | | | | | | | |
| | c | | Homozygous - having the same allele on both chromosomes of a pair e.g. AA or aa Heterozygous - when the alleles on a pair of chromosomes are different e.g. Aa | 2 | DO NOT ALLOW 'have same gene on both chromosomes' DO NOT ALLOW 'have different genes on both chromosomes' | | | | | | | | | |
| | | | Total | 6 | | | | | | | | | | |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance |
|----------|---|----|---|-------|--|
| 4 | a | i | 0.87 | 1 | <p>Examiner's Comments</p> <p>This question on the whole was answered well with many candidates selecting 0.87. The most common incorrect answer was 1 in 87.</p> |
| | | ii | idea that she would definitely develop breast cancer / it would be certain | 1 | <p>accept it is 100% (certain/she will get cancer / she is going to get it) ignore any answer that suggests she already has it, including "she will have it"s unless it is qualified</p> <p>Examiner's Comments</p> <p>Many candidates failed to gain marks on this question. Many thought that Jane either had cancer or had a low chance of getting cancer; very few seemed to realise that it would in fact make it likely that she would get cancer. Few candidates were confident enough to say that she was certain to get cancer. Centres should be encouraged to develop candidates' understanding of probability.</p> |
| | b | | <p>any 3 from:</p> <p>her risk of developing breast cancer is high ;</p> <p>(but) she may not develop breast cancer / it is not certain ;</p> <p>reference to risk/pain/side-effects/scars/death/infection ;</p> | 3 | <p>do not credit unqualified idea that it is major surgery, as this is given in the question</p> <p>ignore 'it may go wrong' ignore 'it may not be safe' ignore ref. to cost accept example of consequence of surgery e.g. she might not be able to breastfeed / body image issues ignore false positive / negative / discrimination / insurance comments / pregnancy</p> |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance |
|----------|--|-------|---|
| | <p>consider how much the surgery would reduce the risk of cancer ;</p> <p>cancer is life-threatening / a very serious disease ;</p> <p>may not eliminate the risk of breast cancer/she might still get breast cancer ;</p> <p>idea that benefits (of surgery) outweigh risk/pain (of surgery) /ORA</p> | | <p>accept 'it may not work'</p> <p>Examiner's Comments</p> <p>This question asked candidates to consider whether Jane should have surgery to remove the breast tissue. Candidates found this question challenging. The majority of candidates scored 1 mark for this question; few candidates scored 3 marks. The responses indicated that the candidates did not understand that this was preventative surgery and not surgery to remove cancerous tissue. Many candidates seemed to think that Jane had cancer and that the surgery was to remove the cancerous tissue. As a result, many incorrectly identified that all the tissue may not be removed and that the cancer may return or discussed a cost element to the surgery. Those that did gain marks did so for correctly identifying that with surgery there are risks or that removing breast tissue could have implications for the future, such as being unable to breast feed or causing body image issues. Very few candidates appreciated that just because the chances of her developing cancer were high this does not mean that she will definitely develop cancer.</p> |

Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|---|--|-------|--|
| | c | <p>any 1 from:</p> <p>could get cancer in other parts of the body/could get other types of cancer ;</p> <p>other factors can cause cancer (e.g. lifestyle/environmental) ;</p> <p>other genes could cause cancer</p> | 1 | <p>do not accept could still be some cancer cells left/ may not have removed/ got rid of all the cancer</p> <p>Examiner's Comments</p> <p>The vast majority of candidates did not answer this question well indicating that they had not understood the question or misinterpreted what the question was asking. Few candidates realised that there were other types of cancer or that lifestyle issues could be involved in causing cancer. Most referred to the 'faulty gene' and that she would be unable to remove this gene hence her risk of cancer would always be present.</p> |
| | d | <p>any 2 from:</p> <p>idea that it is unlikely/rare/low chance/0.1% chance that the normal allele will become faulty ;</p> <p>idea that Jane inherited the faulty allele/it from her mother ;</p> | 2 | <p>do not credit ref. to "1 in 1000" unqualified, as this is given in the question</p> <p>accept the idea that it was passed on ignore unqualified reference to being a carrier</p> |

Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|--|--|----------|---|
| | | idea that Jane's mother inherited the faulty allele/it from Jane's grandmother ; | | <p>ignore "Jane inherited it from her grandmother" / "it skipped a generation" as this does not support the doctor's conclusion</p> <p>Examiner's Comments</p> <p>Candidates found this question challenging. Most answers contained a discussion about who may or may not have had the faulty allele rather than identifying the line of inheritance from grandmother to mother. Candidates did correctly identify that the father did not have the faulty allele and was therefore not responsible; unfortunately, this did not score a mark. There was clear evidence that candidates could use a family tree, but they seemed to struggle to communicate their thoughts. It was rarely mentioned that the normal allele had only a 0.1% chance of becoming faulty despite the question stem pointing candidates to both the family tree and the information about the allele.</p> |
| | | Total | 8 | |
| 5 | | human body cells <div style="display: flex; flex-direction: column; align-items: flex-end; margin-top: 10px;"> <input type="checkbox"/> <input checked="" type="checkbox"/> <input type="checkbox"/> <input type="checkbox"/> </div> | 1 | <p>two or more ticks = 0 marks</p> <p>Examiner's Comments</p> <p>Candidates were asked to identify which cells in a human usually contain pairs of chromosomes. Many candidates selected the correct response. The most common error observed showed that candidates thought that all human cells contained pairs of chromosomes.</p> |
| | | Total | 1 | |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance |
|----------|---|-------|---|
| 8 | <p>[Level 3] Gives a good explanation as to why the baby is different to Poppy AND to its parents. Quality of written communication does not impede communication of the science at this level.</p> <p style="text-align: right;">(5–6 marks)</p> <p>[Level 2] Gives a good explanation as to why the baby is different to Poppy OR to its parents. Quality of written communication partly impedes communication of the science at this level.</p> <p style="text-align: right;">(3–4 marks)</p> <p>[Level 1] Makes any correct statement about the processes involved not linked to Poppy or parents. Quality of written communication impedes communication of the science at this level.</p> <p style="text-align: right;">(1–2 marks)</p> <p>[Level 0] Insufficient or irrelevant science. Answer not worthy of credit.</p> <p style="text-align: right;">(0 marks)</p> | 6 | <p>This question is targeted at grades up to C</p> <p>Indicative scientific points may include:</p> <p>General reasons for differences (applicable to parents and Poppy)</p> <ul style="list-style-type: none"> • variation • inherit different (combinations) of chromosomes • inherit different (combinations) of genes / alleles • alleles can be dominant / recessive • environmental factors <p>Reasons why baby is different to Poppy:</p> <ul style="list-style-type: none"> • sexual reproduction • different egg & sperm • genetic abnormalities / mutation • ref to the fact it could be a boy • different combination of sex chromosomes <p>Reasons why new baby is different to parents:</p> <ul style="list-style-type: none"> • each parent only contributes half the genes / alleles • will show some similarities but won't be identical <p><u>Examiner's Comments</u></p> <p>This was the first of the six-mark extended-writing questions, and many candidates found this level of response question difficult. Candidates were required to give an explanation as to the reasons why a baby born to the same parents would be different to their sister, Poppy, and the child's parents. Many candidates failed to fully address the question and, whilst many candidates correctly identified the reason for these differences would be as a result of different genes, they often found it</p> |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance |
|----------|--|--|--|----------|---|
| | | | | | <p>difficult to express and failed to attach this reason to Poppy or to the parents. This limited the candidates to a Level 1 answer.</p> <p>Candidates were able to give other basic reasons as to why there were differences. These included reference to the role played by the environment and identifying that the baby could have been a different sex to Poppy. Some candidates referred to identical twins, highlighting that this would be the only occasion when the children would be identical. More able candidates introduced the idea of different allele combinations and random gamete fertilisation.</p> <p>Occasionally candidates did not read the information correctly and talked about the new baby having a different mum or dad.</p> |
| | | | Total | 6 | |
| 9 | | | <p>Pair of alleles</p> <div style="display: flex; justify-content: space-around;"> <div style="border: 1px solid black; padding: 5px; width: 150px;">two dominant alleles</div> <div style="border: 1px solid black; padding: 5px; width: 150px;">the associated dominant characteristic</div> </div> <div style="display: flex; justify-content: space-around; margin-top: 10px;"> <div style="border: 1px solid black; padding: 5px; width: 150px;">one dominant and one recessive allele</div> <div style="border: 1px solid black; padding: 5px; width: 150px;">the associated recessive characteristic</div> </div> <div style="display: flex; justify-content: space-around; margin-top: 10px;"> <div style="border: 1px solid black; padding: 5px; width: 150px;">two recessive alleles</div> </div> | | |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance |
|----------|---|----|---------------------------|-------|---|
| 10 | a | | males: XY females: XX (1) | 1 | <p>need both for the mark</p> <p>Examiner's Comments</p> <p>A surprising number of candidates did not score a mark for this question. Common incorrect responses included the use chromosome numbers (23) or single letters (X or Y).</p> |
| | b | i | 1000 (1) | 1 | <p>Examiner's Comments</p> <p>This mathematical question did not appear to cause candidates any problems and the majority were awarded the mark.</p> |
| | | ii | 1200 (2) | 2 | <p>correct answer = 2 marks</p> <p>allow one mark for correct working $1.2 \times 1000 / (0.2 \times 1000) + 1000$</p> <p>Examiner's Comments</p> <p>Candidates found this second mathematical question difficult with few candidates scoring any marks. Common incorrect calculations included adding 1.2 to 1000 or dividing 1000 by 1.2. A higher number of candidates did not attempt this question compared to other questions.</p> |

Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|-----|--|----------|---|
| | iii | female fetuses terminated (1) female fetuses miscarried (1) disease affecting females fetuses (1) X sperm killed / more Y sperm (1) gender selection (1) | 2 | do not allow 'kill female baby' do not allow 'more boy sperm' 'female fetuses aborted because they want boys' = 2 marks Examiner's Comments This also proved to be a difficult question. Those candidates that did score 2 marks were often awarded the marks for identifying that the ratio could have been a result of the termination of female foetuses. Those candidates gaining one mark frequently did so for reference to gender selection. Many candidates were familiar with China's one child policy but did not always apply this knowledge effectively. Incorrect responses included suggestions that the male chromosome/gene may be stronger. |
| | | Total | 6 | |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance | | | | | | | | | | | | | | | |
|----------|---|--------|--|-------|--|--------|--|--|--|---|---|------|---|----|----|---|----|----|---|--|
| 11 | a | i | <div style="text-align: center;"> <table border="1" style="margin: auto;"> <tr> <td colspan="2"></td> <td colspan="2" style="text-align: center;">Sharon</td> </tr> <tr> <td colspan="2"></td> <td style="text-align: center;">T</td> <td style="text-align: center;">t</td> </tr> <tr> <td rowspan="2" style="text-align: center;">Eric</td> <td style="text-align: center;">T</td> <td style="text-align: center;">TT</td> <td style="text-align: center;">Tt</td> </tr> <tr> <td style="text-align: center;">t</td> <td style="text-align: center;">Tt</td> <td style="text-align: center;">tt</td> </tr> </table> <p style="text-align: right;">(2)</p> </div> | | | Sharon | | | | T | t | Eric | T | TT | Tt | t | Tt | tt | 2 | <p>one mark for correct parent genotypes (both Tt)</p> <p>one mark for correct completion of Punnett Square</p> <p>ecf for correct completion of Punnett Square from their genotypes</p> <p>allow tT for Tt</p> <p>Examiner's Comments</p> <p>Most candidates successfully completed the Punnett square and gained both marks for this question. Centres should remind candidates to take care with the letters used to complete the Punnett square and ensure that the difference between the lower case letter and upper case letter is clear. There were occasions when this difference was not clear and resulted in marks being lost. Examiners were instructed to use an error carried forward for the second marking point to ensure candidates were not penalised for the same mistake twice.</p> |
| | | Sharon | | | | | | | | | | | | | | | | | | |
| | | T | t | | | | | | | | | | | | | | | | | |
| Eric | T | TT | Tt | | | | | | | | | | | | | | | | | |
| | t | Tt | tt | | | | | | | | | | | | | | | | | |
| | | ii | 0.25 / 1/4 / 25% / 1 in 4 / 1:3 | 1 | <p>ecf from Punnett Square i.e. this probability must match their Punnett Square in (b)(i)</p> <p>ignore 1 in 3 do not allow 3:1 / 4:1 / 1:4</p> <p>Examiner's Comments</p> <p>Candidates found this question particularly difficult even if marks had been awarded in part (b) (i). There appeared to be a misunderstanding as to the genotype which would give rise to cystic fibrosis with many candidates incorrectly identifying Tt. As a result common incorrect answers were 75% and 50%.</p> | | | | | | | | | | | | | | | |

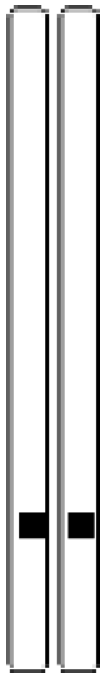
Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|---|--|----------|---|
| | b | <p>identify a positive reason for testing (1)</p> <p>(idea that) the benefits outweigh the risk (1)</p> | 2 | <p>allow idea of planning (even though given in the question)</p> <p>do not allow the idea that they want to know if the fetus has the disease unqualified</p> <p>allow inference that benefit outweighs the risk, e.g. "even though the test may be painful and inaccurate, it is more important to find out if the fetus has cystic fibrosis"</p> <p>note that mark-points may be linked, e.g. "benefits of being able to plan outweigh the possible problems" = 2 marks</p> <p>Examiner's Comments</p> <p>The vast majority of candidates scored one mark for this question more often than not for the identification that a benefit would be to plan treatment for the baby. Many candidates stated a benefit and a risk but did not develop this further to identify that the benefits outweigh the risks and therefore did not gain the second mark.</p> |
| | | Total | 5 | |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance |
|----------|---|----------|---|
| 12 | <p>Level 3 (5–6 marks) Good explanation of both Huntington's and cystic fibrosis using dominant and recessive with genotypes and correct and relevant diagrams used in explanation. Plus additional information such as explanation of carriers and chances of inheritance.</p> <p>Quality of written communication does not impede communication of the science at this level.</p> <p>Level 2 (3–4 marks) Correct explanation of Huntington's or cystic fibrosis using dominant and recessive. Quality of written communication partially impedes communication of the science at this level.</p> <p>Level 1 (1–2 marks) Makes a simple statement that Huntington's is dominant or cystic fibrosis is recessive.</p> <p>Quality of written communication impedes communication of the science at this level.</p> <p>Level 0 (0 marks) Insufficient or irrelevant science. Answer not worthy of credit.</p> | 6 | <p>This question is targeted at grades up to C</p> <p>Indicative scientific points at Level 3 may include</p> <ul style="list-style-type: none"> • correct diagram used to assist explanation • both cystic fibrosis and Huntington's correctly explained <p>Indicative scientific points at Level 2 may include</p> <ul style="list-style-type: none"> • cystic fibrosis – correct idea that can be carrier • both recessive alleles must be present to be a sufferer • Huntington's – only 1 allele means sufferer • diagram may be missing or incorrect <p>Indicative scientific points at Level 1 may include</p> <ul style="list-style-type: none"> • Cystic fibrosis is recessive • Huntington's is dominant • some linking of cystic fibrosis / carrier <p>Use the L1, L2, L3 annotations in Scoris; do not use ticks.</p> <p>Examiner's Comments</p> <p>Candidates were asked to explain how cystic fibrosis and Huntington' s disease are inherited. The stem of the question asked candidates to explain this, using genetic diagrams. Many candidates knew that cystic fibrosis was recessive and that Huntingdon' s was dominant. Few were able to use genetic diagrams effectively to explain the inheritance and many failed to use genetic diagrams in their answers, limiting the number of marks they could score.</p> |
| | Total | 6 | |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance |
|----------|--|----|---|----------|---|
| 13 | | i |  | 1 | <p>gene in identical position to original give mark as long as there is a 50% overlap</p> <p>Examiner's Comments</p> <p>It was encouraging to see that more than half of the candidates were able to draw the second gene in the correct position.</p> |
| | | ii | 2 | 1 | <p>accept any indication of correct response – eg underlining etc.</p> <p>Examiner's Comments</p> <p>In this question candidates were asked to circle the maximum number of alleles. Candidates who failed to score here tended to circle 46 or 23 as the correct answer.</p> |
| | | | Total | 2 | |

Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|--|---|----------|--|
| 14 | | any 3 from: conclusion 1: 50:50 or 50% chance or probability ; idea that it could be slightly more or less / will not be exactly 50/50; conclusion 2: is wrong; (having 2 girls) does not change probability / likelihood / still 50% (chance) | 3 | could state that conclusion 1 is correct or incorrect answer which states that 50 will be male does not score this mark could cover both conclusions together without specifically relating to 1 or 2 by stating that both are wrong. Examiner's Comments This question required candidates to comment on the conclusions having applied their knowledge of the inheritance of gender to the conclusions given. Most candidates who scored on this question knew that the statements were incorrect; however, few were able to explain why. |
| | | Total | 3 | |

Mark Scheme

| Question | | | Answer/Indicative content | Marks | Guidance |
|----------|--|---|---------------------------|------------|---|
| 15 | | i | TT ✓ | 1 (AO 2.1) | <p>more than one tick = 0 marks</p> <p><u>Examiner's Comments</u></p> <p>This AO2 question was attempted by all candidates, with two thirds selecting the correct answer indicating that they knew the meaning of the term homozygous.</p> |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance | | | | | | | | | | | | | | | | | | | | | | | | | | | |
|----------|--|-------|----------|-----|---|----|----|---|----|------|----------------|---|--|---|-----|---|----|----|---|----|------|--|---|-----|---|----|----|---|----|------|
| ii | <table border="1" style="margin: auto;"> <tr><td></td><td>T</td><td>T ✓</td></tr> <tr><td>t</td><td>Tt</td><td>Tt</td></tr> <tr><td>t</td><td>Tt</td><td>Tt ✓</td></tr> </table> | | T | T ✓ | t | Tt | Tt | t | Tt | Tt ✓ | 2 (AO 2.2 × 2) | <p>one mark awarded for correct gamete genotypes</p> <p>one mark awarded for correct offspring genotypes derived from gamete genotypes</p> <p>ALLOW ECF from (a)(i)</p> <p>if Tt or tT identified as genotype</p> <div style="text-align: center;"> <table border="1" style="margin: auto;"> <tr><td></td><td>T</td><td>t ✓</td></tr> <tr><td>t</td><td>Tt</td><td>tt</td></tr> <tr><td>t</td><td>Tt</td><td>tt ✓</td></tr> </table> </div> <p>or if tt identified as genotype</p> <div style="text-align: center;"> <table border="1" style="margin: auto;"> <tr><td></td><td>t</td><td>t ✓</td></tr> <tr><td>t</td><td>tt</td><td>tt</td></tr> <tr><td>t</td><td>tt</td><td>tt ✓</td></tr> </table> </div> <p>Examiner's Comments</p> <p>Candidates were not penalised if they did not correctly identify Jack's genotype, as the punnet square was marked using the genotype identified in 3(a)(i). Many candidates were successfully able to complete the punnet square, although not all of them were then able to correctly derive the probability based on their genetic cross. This question assessed objectives AO2 and AO3.</p> | | T | t ✓ | t | Tt | tt | t | Tt | tt ✓ | | t | t ✓ | t | tt | tt | t | tt | tt ✓ |
| | T | T ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | Tt | Tt | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | Tt | Tt ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| | T | t ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | Tt | tt | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | Tt | tt ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| | t | t ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | tt | tt | | | | | | | | | | | | | | | | | | | | | | | | | | | | |
| t | tt | tt ✓ | | | | | | | | | | | | | | | | | | | | | | | | | | | | |

Mark Scheme

| Question | | Answer/Indicative content | Marks | Guidance |
|----------|-----|---|----------------|---|
| | iii | 1 / 100% / certain ✓ | 1 (AO 3.2a) | <p>ALLOW ECF from (a) (ii)</p> <p>if Tt or tT identified as genotype probability is 0.5 / 50% / ½ ✓</p> <p>if tt identified as genotype probability is 0% ✓</p> |
| | iv | <p>Any two from:</p> <p>sex chromosomes/ X and Y chromosomes / 23rd pair of chromosomes ✓</p> <p>male is XY and female is XX ✓</p> <p>genes on the Y chromosome trigger the development of testes ✓</p> <p>50% sperm carry X and 50% carry Y so the outcome of XY and XX is 50:50 ✓</p> | 2 (AO 1.1 × 2) | <p>ALLOW female has XX chromosomes and male has XY chromosomes for 2 marks</p> <p><u>Examiner's Comments</u></p> <p>This AO1 question assessed knowledge in isolation. Candidates who achieved lower overall marks did not understand what they were being asked to do in this question. Some thought it was asking about the physical features which determine a male or a female. A significant number of candidates thought that this question related to the choices individuals make in personal relationships.</p> |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance |
|----------|--|----------------|---|
| v | <p>contraceptive pill ✓ <i>plus one from:</i> because it prevents ovulation / prevents release of an ovum or egg ✓</p> <p>thickens the mucus of the cervix so sperm can't pass through ✓</p> <p>if they are happy to have unprotected sex / AW ✓</p> <p>OR condom ✓ <i>plus one from:</i> because it prevents sperm reaching ovum or egg / prevents sperm entering the vagina or cervix or uterus ✓</p> <p>also protects against spread of STIs / AW ✓</p> <p>OR female condom ✓ <i>plus one from:</i> because it prevents sperm reaching ovum or egg / prevents sperm passing into the uterus or through the cervix ✓</p> <p>also protects against spread of STIs / AW ✓</p> <p>OR intra-uterine device / system or IUD or coil ✓ <i>plus one from:</i> can remain in place for a long time or up to ten years ✓</p> <p>prevents sperm surviving in the uterus ✓</p> <p>stops egg or ovum being fertilised ✓</p> <p>prevents embryo implanting in the uterus ✓</p> <p>OR diaphragm ✓ <i>plus</i> prevents sperm entering the uterus ✓</p> | 2 (AO 1.1 × 2) | <p>suggested form of contraception = 1 mark justification = 1 mark</p> <p>IGNORE for condom, IUD and diaphragm justification that refers to sperm not entering the woman's body</p> <p>IGNORE throughout justification referring</p> |

Mark Scheme

| Question | Answer/Indicative content | Marks | Guidance |
|----------|---|----------|---|
| | <p>OR contraceptive implant ✓ <i>plus one from:</i> works for up to 3 years ✓</p> <p>prevents ovulation / prevents ovum or egg being released ✓</p> <p>OR surgical method / vasectomy / sterilisation ✓ <i>plus one from:</i> a permanent solution (as they don't want any more children) ✓</p> <p>prevents eggs and sperm meeting as tubes are cut ✓</p> <p>no sperm released with vasectomy ✓</p> <p>egg cannot pass down oviduct with female sterilisation ✓</p> <p>OR rhythm method / abstinence ✓ <i>plus one from:</i> no chemicals are used ✓</p> <p>there are no religious or ethical objections ✓</p> | | <p>to cost or availability or efficacy or safety</p> <p>DO NOT ALLOW prevents eggs or sperm being made</p> <p>Examiner's Comments</p> <p>A large number of candidates could suggest a method of contraception using correct biological terminology. However, they found it more difficult to justify their choice using biological reasons. Many said simply the method would stop Nina becoming pregnant, or referred to the availability, cost or percentage efficacy of the method chosen. This question assessed objective AO1.</p> |
| | Total | 8 | |