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| *Human Perspectives ATAR Units 1 & 2* |

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Answers

Chapter 15 Genetics can be used to understand the traits of individuals and families

Questions 15.1

Recall knowledge

**1** What is another term for trait?

Answer*:* Characteristic

**2** What organisms were used in Mendel’s experiments?

Answer*:* Pea plants

**3** Describe the difference between pure-breeding and hybrid.

Answer*:* Pure-breeding is when organisms produce the same characteristic in each succeeding generation when bred among themselves. A hybrid is the result of crossing two pure-breeding organisms of contrasting traits. They contain the genetic information of both traits, although would show only one of them.

**4** Define:

**a** dominant trait

Answer*:* One of a pair of contrasting characteristics, which is controlled by an allele that is not masked by any other alleles. The characteristic shown by the hybrid.

**b** recessive trait

Answer*:* One of a pair of contrasting characteristics, which is controlled by an allele that is masked by dominant alleles. The characteristic not shown in the hybrid.

**5** Describe the principle of segregation.

Answer*:* The principle of segregation occurs during meiosis, where each resulting gamete receives only one gene for each trait.

**6** Describe the difference between:

**a** homozygous and heterozygous

Answer*:* Homozygous has the same alleles for the trait, seen in pure-breeding organisms. Heterozygous is seen in the hybrid, where the hybrid contains different alleles for the same trait.

**b** genotype and phenotype.

Answer*:* The genotype is the allele combinations, represented by letters eg: *aa* or *Gg*. The phenotype is the physical appearance of the organism determined by the genetic combination e.g. tall or green.

Apply knowledge

**7** Explain why it was important that Mendel’s work matched the observations of Sutton’s work on cells.

Answer*:* Sutton’s observations were on the behaviour of chromosomes during meiosis and the suggestion that hereditary factors (now known as genes) were located on the chromosomes. This linked to Mendel’s speculation that the chromosomes split during meiosis, with each gamete receiving one of the chromosome pair and when cross-breeding occurred, the resulting hybrid would show one of the traits of the parents. This led to the chromosome theory of heredity.

**8** Explain why it was important that the pea plants originally used by Mendel were pure-bred.

Answer*:* They needed to be pure-bred to ensure there was no other factor that could influence the outcome of the hybrid cross.

**9** Explain why the letter *x* is not a suitable choice to represent an allele.

Answer*:* The letter x is used as the symbol to show the cross between two parents. If the alleles were also using the letter x there could be confusion as to the genotypes of the parents.

**10** Classify each of the following genotypes as homozygous or heterozygous:

**a** Rr

Answer*:* Heterozygous

**b** GG

Answer*:* Homozygous

**c** Tt

Answer*:* Heterozygous

**d** aa

Answer*:* Homozygous

**e** EE

Answer*:* Homozygous

**11** State whether each of the following is a genotype or phenotype.

**a** curly hair

Answer*:* Phenotype

**b** *Rr*

Answer*:* Genotype

**c** *HH*

Answer*:* Genotype

**d** blue eyes.

Answer*:* Phenotype

**12** In Mendel’s experiments, there were no short plants in the first filial generation. However, approximately one-quarter of the plants in the second filial generation were short. Explain this observation.

Answer*:* One parent for the first cross was homozygous for tall and the other was homozygous for short. The offspring produced were all hybrids, or heterozygotes, containing an allele for tall and short in their genotype. However, tall is a dominant trait over short, so phenotypically all the offspring of the first filial generation were tall. In crossing the first filial generation (both parents were heterozygous), 25% of the offspring received a short allele from each parent. As such, genotypically they were homozygous for short, and phenotypically showed the short trait.

Questions 15.2

Recall knowledge

**1** The probability of a child having a trait is 25%. What is this probability as a fraction?

Answer: ¼

**2** How are each of the following represented in a pedigree?

**a** Male with the trait

Answer*:* A shaded-in square

**b** A deceased female with the trait

Answer*:* A shaded-in circle with a diagonal line through it

**c** Children from the same parents

Answer*:* Circles or squares branching off from the same line linked to the parents in the generation above.

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**d** The generation

Answer*:* Roman numerals on the left-hand side of the diagram

**e** A female without the trait

Answer*:* An unshaded circle

**3** Define ‘consanguineous’.

Answer*:* A union between the two close relatives, usually cousins, represented by double horizontal lines connecting the individuals.

Apply knowledge

**4** Suggest a genotype for someone with no eye folds.

Answer*:* Homozygous recessive eg: *ee*

**5** Draw a Punnett square for each of the following crosses.

**a** RR × rr

Answer:

|  |  |  |
| --- | --- | --- |
|  | *R* | *R* |
| *r* | *Rr* | *Rr* |
| *r* | *Rr* | *Rr* |

**b** Mm × Mm

Answer:

|  |  |  |
| --- | --- | --- |
|  | *M* | *m* |
| *M* | *MM* | *Mm* |
| *m* | *Mm* | *mm* |

**c** Ee × ee

Answer:

|  |  |  |
| --- | --- | --- |
|  | *E* | *e* |
| *e* | *Ee* | *ee* |
| *e* | *Ee* | *ee* |

**6** What is the probability that a child will have astigmatism if neither of his parents have the condition?Explain your answer.

Answer*:* There is no chance the child will have astigmatism. Astigmatism is a dominant trait; if the child’s parents have normal vision, they are both homozygous recessive (aa), so there is no dominant allele present in either parent to pass onto their child.

**7** Two parents with broad lips have a child with thin lips. Use a Punnett square to determine the genotype of the parents.

Answer:

Key: B = Broad lips, b = thin lips

Bb x Bb

|  |  |  |
| --- | --- | --- |
|  | *B* | *b* |
| *B* | *BB* | *Bb* |
| *b* | *Bb* | *bb* |

Parents must be heterozygote, if they are to provide their child with the two recessive alleles required to show thin lips.

**8** Use the pedigree below to answer the following questions.



**a** How many females have the trait?

Answer: Three

**b** If the trait is dominant, what is the genotype of the male in generation I?

Answer*:* Male must be homozygous recessive eg: aa

**c** If the trait is recessive, what is the genotype of the:

**i** female in generation II?

Answer*:* Female must be homozygous recessive eg: aa

**ii** unaffected females in generation III?

Answer*:* Females can only be heterozygous eg: Aa

Questions 15.3

Recall knowledge

**1** List five single-gene disorders.

Answer:

* Huntington’s disease
* Achondroplasia
* Facioscapulohumeral muscular dystrophy
* Neurofibromatosis
* Cystic fibrosis
* Albinism
* Sickle-cell anaemia
* Tay Sachs
* Phenylketonuria

Other examples may be acceptable, provided they are inherited by a single gene.

**2** Describe the symptoms of Huntington’s disease and phenylketonuria.

Answer: Huntington’s disease: occasional flailing movement of arms and legs, difficulty in making voluntary movements, writhing movements of hands, head, trunk and feet, dementia.

Phenylketonuria: the build-up of phenylalanine leads to damage to a growing brain, extreme intellectual disability, tendency towards epileptic seizures and a failure to produce normal skin pigmentation.

**3** Describe how someone can be a carrier of a genetic disorder.

Answer*:* A carrier is a person who has a heterozygous genotype; they carry the recessive allele in their genes but do not show the recessive phenotype. They would have received the recessive allele from one of their parents.

**4** Name the test used to screen for phenylketonuria.

Answer*:* A heel prick is done to screen for phenylketonuria.

Apply knowledge

**5** Explain why disorders due to a recessive allele are passed on more frequently than those due to a dominant allele.

Answer*:* The recessive allele is masked by the dominant allele and not seen in the phenotype, so people can be carriers of the disorder and may never know they have the recessive allele. As a result, the recessive allele can be passed on to the offspring more frequently than a dominant disorder that is not masked and is seen in the phenotype of the individual who has it.

**6** Discuss the use of genetic screening with regards to Huntington’s disease. Include possible reasons people may choose, or not choose, to have screening done.

Answer*:* Genetic screening is available for people who has a parent with Huntington’s disease or has died from it. This allows the close relatives an opportunity to know if they have inherited the allele, as the symptoms do not become apparent until after a person is 40 years old. People may choose to have the screening done if they wish to have children and are concerned they might pass the allele on to them. A person may not want to have the screening done because there is no cure for the condition, and they cannot undo the knowledge they have a fatal disease if they receive a positive result to Huntington’s disease,

**7** Explain why it is possible to be a carrier of a recessive disorder, but not a dominant one.

Answer*:* A carrier is a person with a heterozygote genotype, and the recessive allele is masked by the presence of the dominant allele. You cannot carry a dominant allele, as it will always be expressed in the phenotype, it is unable to be masked by other alleles.

**8** State whether the pedigree below is for a recessive or dominant disorder. Justify your answer.



Answer:

This pedigree is for a recessive disorder. Parents in Generation I do not show the disorder but produce an affected child in generation II. This indicates that both parents are carriers and had a 25% chance of producing an affected offspring.

Questions 15.4

Recall knowledge

**1** State the number of:

**a** autosomal chromosomes in a somatic cell

Answer*:* 44 single chromosomes or 22 pairs.

**b** sex chromosomes in a human somatic cell

Answer*:* 2 single chromosomes or one pair of sex chromosomes; in females they are matching, in males they are not.

**c** autosomal chromosomes in a human gamete

Answer*:* 22 single chromosomes.

**d** sex chromosomes in a human gamete

Answer*:* 1 single chromosome.

**e** homologous pairs in a human somatic cell.

Answer*:* In a female there are 23 pairs of homologous chromosomes; in a male there are 22 pairs of homologous chromosomes (the sex chromosomes are not a matching pair).

**2** Draw a Punnett square to confirm that there isa 50% chance of a child being male.

Answer:

Key: XX = female, XY = male

XX x XY

|  |  |  |
| --- | --- | --- |
|  | *X* | *X* |
| *X* | *XX* | *XX* |
| *Y* | *XY* | *XY* |

**3** State the sex chromosomes of a:

**a** female

Answer: XX

**b** male.

Answer: XY

**4** Is the *X* or *Y* chromosome longer?

Answer*:* The *X* chromosome is longer than the *Y* chromosome.

**5** Describe the symptoms of red–green colour blindness.

Answer*:* The individual is unable to distinguish between the colours of red and green because the colours look too similar.

**6** Explain why males are referred to as ‘hemizygous’ when referring to sex-linked characteristics.

Answer*:* Males only possess one *X* and one *Y* chromosome, there is no allelic counterpart for males, so they cannot be referred to as homozygous or heterozygous with respect to sex-linked traits.

Apply knowledge

**7** It could be said that the father determines the sex of the child. Discuss whether this statement is true.

Answer*:* Males are able to provide half their gametes with an *X* chromosome and the other half with a *Y* chromosome. Females are only able to provide an *X* chromosome to all their gametes. So, yes, this statement is true, in that the males are the only source of sex chromosome variation in gametes to determine the sex of their children.

**8** Explain why sex-linked characteristics have an allele on the *X* chromosome but not on the *Y* chromosome.

Answer*:* The *Y* chromosome is not homologous to the *X* chromosome – it contains different genes. Sex-linked characteristics are also called X-linked characteristics because their alleles are only found on the *X* chromosome.

**9** A colour-blind father has a child with a female with normal vision. What is the chance of them having:

**a** a child with normal vision?

Answer*:* This is dependent of the mother’s genotype. If the mother is homozygous dominant, then 100% of the offspring from the union will have normal vision irrespective of their sex.

If the mother is a carrier (heterozygous) then 50% of the females will have colour blindness and 50% of the males will have colour blindness.

**b** a colour-blind daughter?

Answer*:* This is dependent on the mother’s genotype.

If the mother is homozygous dominant, there is 0% chance of a colour-blind daughter. If the mother is a carrier (heterozygous) then 50% of the females will have colour blindness.

**c** a son with normal vision?

Answer*:* This is dependent on the mother’s genotype.

If the mother is homozygous dominant, then there is a 100% chance the son will have normal vision. If the mother is a carrier (heterozygous) then there is a 50% chance the son will have colour blindness.

**10** Explain why haemophilia is much more common in males than in females.

Answer*:* Males only inherit one *X* chromosome – there is no allelic counterpart on the *Y* chromosome. If a male inherits the recessive allele for haemophilia on his *X* chromosome, he will have the disease. There is no way to mask a single recessive allele in males inherited on the *X* chromosome.

**11** If a daughter has a sex-linked disorder, her father must also have it. Justify this observation.

Answer*:* A daughter must have the sex chromosome pair of XX. One X has come from her mother, and the other has come from her father. For a female to show a sex-linked disorder, she must be homozygous recessive and have inherited the two alleles from both her parents. As males only have one X chromosome, in this instance, that chromosome must have the recessive allele on it, and therefore the father would also have the disease.

Questions 15.5

Recall knowledge

**1** Describe co-dominant inheritance.

Answer*:* Co-dominance is a situation where two or more alleles are equally dominant. If both alleles are present in a genotype, they are equally represented in the phenotype.

**2** The alleles for the ABO blood group are represented by *IA*, *IB* and *i*. Explain why:

**a** the letter *i* is used for all alleles

Answer*:* The letter I stands for isoagglutinogen, or antigen. This refers to the alleles that are responsible for the presence of antigens on the surface of red blood cells.

**b** two alleles use a capital *I* but the other is lower case

Answer*:* The capital I is used to show the dominant alleles of A and B, as *IA* and *IB* are co-dominant alleles. The lowercase *i* is used to show the recessive allele.

**c** the letters *A* and *B* are used.

Answer*:* The letters *A* and *B* are used to represent the alleles that produce the antigen A and the antigen B.

**3** State the genotype for someone with type MN blood.

Answer: BMBN

**4** What is the blood type of someone with an *IAi* genotype?

Answer*:* Their blood type would be A.

Apply knowledge

**5** Justify the ABO blood group being an example of both co-dominance and multiple allele inheritance.

Answer*:* The ABO blood group is an example of co-dominance as the alleles *IA* and *IB* are equally represented in the heterozygote individual (their blood type would be AB and both antigen A and antigen B are present on their red blood cells). It is also an example of multiple alleles, as the genes for these antigens are located at the same location on chromosome 9.

**6** A child’s blood type is AB. List all the possible genotypes of the parents, including Punnett squares to support your answers.

Answer:

The child’s genotype has to be IAIB

Parents can be IAIA xIBIB and will have a 50% chance of a child with AB blood type.

|  |  |  |
| --- | --- | --- |
|  | *IA* | *IA* |
| *IB* | *IAIB*  | *IAIB*  |
| *IB* | *IAIB*  | *IAIB*  |

Parents can be *IAi*x*IBi* and will have a 25% chance of a child with AB blood type.

|  |  |  |
| --- | --- | --- |
|  | *IA* | *i* |
| *IB* | *IAIB*  | *IAi* |
| *i* | *IAi* | *ii* |

Parents can be *IAi*x*IAIB* and will have a 25% chance of a child with AB blood type.

|  |  |  |
| --- | --- | --- |
|  | *IA* | *i* |
| *IA* | *IAIA*  | *IAi* |
| *IB* | *IAIB* | *IBi* |

Parents can be *IBi*x*IAIB* and will have a 25% chance of a child with AB blood type.

|  |  |  |
| --- | --- | --- |
|  | *IB* | *i* |
| *IA* | *IAIB*  | *IAi* |
| *IB* | *IBIB* | *IBi* |

**7** Explain why a parent with type O blood can have a child with type A blood, but not type AB.

Answer*:*  A parent with type O blood has the genotype of *ii*. If the other parent has the phenotype of A, they can have either a genotype of *IAIA* or *IAi*.

*IAIA*× ii  will result in 100% offspring *IAi* Genotype and 100% chance of blood type A.

*IAi* × ii will result in 50% offspring *IAi* Genotype, and 50% chance of blood type A.

Questions 15.6

Recall knowledge

**1** Define ‘genetic counselling’.

Answer*:*  Genetic counselling is the advice given to prospective parents after the analysis of the incidence of a disorder in their family trees. The probability that a particular disorder may be inherited is discussed, and the couple are given all the information to make a choice about the risk of having a baby with the inherited disorder.

**2** Name the technique used to determine a DNA fingerprint.

Answer*:* Electrophoresis

**3** List some situations where genetic profiling could be considered to be:

**a** a positive option

Answer*:*  Future diseases may be predicted so early intervention and early treatment may be possible.

Knowing your genetic makeup helps make a more informed choice about having children that can inherit the same genetic disease.

**b** a negative option.

Answer: Knowing you have a genetic disease that has no treatment or prevention may lead to unnecessary anxiety and can change a person’s perception of wellness.

In determining paternity, cases of divorce have arisen as fathers discover they are raising a child that is not biologically their own.

Discrimination where genetic information could be used to increase health insurance premiums, or life insurance companies can demand genetic profiling before agreeing to supply cover.

Employers may discriminate against potential employees with a particular genetic profile.

Apply knowledge

**4** Explain why some couples may choose to seek genetic counselling prior to starting a family.

Answer*:*  Either or both individuals may have a family history of genetic diseases, or in the case of an adoption, may not be aware of any family history. Genetic screening will provide the information required to make a more informed choice about having children.

**5** DNA profiling can be used to identify paternity. Use the DNA profiles below to identify which male isthe father of the child.



Answer*:*  Male 1 is the father. The child has 5 bands from the mother, and the remaining 6 bands match with Male 1.

**6** Explain why 20–40 markers are studied in DNA profiling, rather than just one.

Answer*:* Specific markers on a DNA strand are studied and the number of repeats at each marker are compared. If only one marker is examined there is a one in ten million chance that two people have the same number of repeats. If 20 markers are examined there is a 1 in 500 000 chance that two people will have the same markers. 40 markers will result in a one in 250 000 chance of two people having the same markers. The more markers that match, the more likely the two individuals are related.

Chapter 15 activities

Activity 15.1 Investigating Mendelian genetic principles in Martians

Studying your data

**1** Because the first three columns all contain the dominant allele (*R*), individuals with these genotypes will all appear red. Tally up the number of red offspring.

Answer*:*  Students results will vary

**2** Individuals with the genotype *rr* will appear white. How many white offspring do you have?

Answer*:*  Students results will vary

**3** What is the ratio of the phenotypes, red to white?

Answer*:*  The ratio should be close to 3:1

**4** Combine your data with that of the other groups in the class to obtain a bigger sample. What is the ratio now?

Answer*:*  The ratio should be close to 3:1

Interpreting your data

**5** Has this activity shown that inheritance of skin colour in Martians follows the principles of Mendelian genetics?

Answer*:* You would expect answers in the affirmative.

**6** How close were your results to the expected result of 3:1?

Answer*:* Answers will vary from group to group.

**7** Was the ratio calculated by combining all groups in the class closer to the expected? Explain why this was the case.

Answer*:* Answers will vary, but one would assume the class result would be closer to the expected ratio.

This is due to the larger sample size for the whole class compared with a single group.

Activity 15.2 Examining pedigrees

Pedigree 1

Interpreting your results

**1** Can you be absolutely certain about the genotypes of all individuals in the first generation? Give reasons for your answer.

Answer*:* Generation I 1 and 2 must be heterozygotes due to Generation II individuals 2 and 3 being Rh- and 5 being Rh+. Generation I 4 is homozygous recessive. Generation I 3 can be either homozygous dominant or heterozygote; there is no way to tell for certain as both their children Generation II 8 & 9 are Rh+.

**2** What are the genotypes of the male (III 5) and the female (III 6) in the third generation who married and produced two daughters? Describe the genotypes using both words and the appropriate letters. Explain why you can be certain of their genotypes.

Answer*:* The male (III 5) must be a heterozygote (Rr).

The female (III 6) must be homozygote recessive (rr).

This is because their daughter Generation IV 4 is homozygote recessive, so must have received a recessive allele from each parent. And Generation IV 5 is heterozygote, having received the dominant allele from her father and recessive allele from her mother.

**3** Do the two Rh+ females in the fourth generation (IV 5 and IV 8) have the same genotype? Explain your answer.

Answer*:* Yes, they would both be heterozygotes (Rr). This is due to them both having a Rh- mother who would be homozygote recessive (rr). As neither of them show the trait, they must also have one dominant allele from their respective fathers.

**4** Is the Rh blood group controlled by a gene on an autosomal or an *X* chromosome?

Answer*:* The Rh blood group gene is located on an autosomal chromosome. In Generation III individual 5 is an unaffected father, who has produced an affected daughter Generation IV 4. If this condition was carried on the *X* chromosome, that father would also be affected. There are also more females affected in this pedigree than males.

Pedigree 2

What to do

Carefully examine the pedigree and work out the genotypes of all individuals shown. Is there any member of the family about whose genotype you are uncertain?

Answer: Key: A= astigmatism, a=normal vision

Gen I 1:AA or Aa 2: aa

Gen II 1:Aa 2: Aa 3: Aa 4:aa 5:Aa 6:aa

Gen III 1:AA or Aa 2: aa 3: Aa 4:aa 5:aa 6:aa 7:Aa 8:aa 9:aa 10:aa 11:aa 12: Aa

Gen IV 1:Aa 2: Aa 3: Aa 4:aa 5:aa 6:aa 7:aa 8:aa 9:Aa 10:aa 11:aa 12:aa 13:Aa 14:Aa

Gen I 1 and Gen III 1 could be either homozygote doinant or heterozygote for astigmatism.

Interpreting your results

Is astigmatism in the family in pedigree above dominant or recessive? Give reasons for your answer.

Answer*:* Astigmatism is a dominant trait. Generation II individuals 1 and 2 have the trait, however they produce 3 offspring Generation III 2, 4 and 5 that do not have the trait. This can only occur when the parents are heterozygote for their genotype. The trait also appears in every generation.

Activity 15.3 Studying a family with Huntington’s disease

What to do

Construct a pedigree to show all the individuals in the family. Indicate the individuals who have Huntington’s disease by shading the relevant circles or squares.

Answer:



**Interpreting the family tree**

**1** Write down the possible genotypes of James, Anne, Jennifer and John. Explain the symbols you are using.

Answer*:* Where *H* represents the allele for Huntington’s disease and *h* represents the allele for the absence of Huntington’s disease:

• James: *Hh*

• Anne: *hh*

• Jennifer: *Hh*

• John: *hh*

**2** What is the probability that Michele has inherited Huntington’s disease? Using a Punnett square, set out the cross between Michele’s parents in full.

Answer:

Hh × hh.

|  |  |  |
| --- | --- | --- |
|  |  | Mother (Jennifer) |
|  |  | *H* | *h* |
| Father (John) | *h* | *Hh* | *Hh* |
| *h* | *Hh* | *hh* |

Genotypes: 50% *Hh* and 50% *hh*

Phenotypes: 50% Huntington’s disease and 50% normal

Therefore, Michele has a 50% chance of inheriting the disease.

**3** Is there any possibility that Darren has inherited the disease? Explain, using a Punnett square to set out the cross between his parents in full.

Answer*:* Michele has a 50% chance of inheriting the disease, and if she does, Darren has a 50% chance of having the disease.

If Michele has the disease, then: *Hh* × *hh*.

|  |  |  |
| --- | --- | --- |
|  | *H* | *h* |
| *h* | *Hh* | *hh* |
| *h* | *Hh* | *hh* |

Genotypes: 50% *Hh* and 50% *hh*

Phenotypes: 50% Huntington’s disease and 50% normal

Therefore, Michele has a 50% chance of having Huntington’s disease. If she does, Darren will have a 50% chance of also having the disease.

Thus, 50% × 50% = 25% chance Darren will have the disease.

**4** Is the gene that determines whether a person has Huntington’s disease located on an autosomal or an *X* chromosome? Explain your answer.

Answer*:* Huntington’s disease is determined by a gene on an autosomal chromosome.

Activity 15.4 Investigating patterns of inheritance in heterozygous barley seeds

Discussion

**1** What is one limitation of this investigation?

Answer*:* The sample size of the barley seeds is one limitation in this procedure. Students may suggest the growing conditions as another.

**2** How would you improve the reliability and validity of the data in this procedure?

Answer*:* A larger sample size and more controlled growing conditions would improve the reliability and validity of the data.

**3** Was your predicted ratio based on your Punnett square proven correct?

Answer*:* Student answers will vary.

**4** Were there any inconsistencies in the results? If so, explain why they may have occurred.

Answer*:* Statistics would suggest a 3:1 ratio, however, a perfect 3:1 ratio is unlikely in real life. Statistical outliers such as all green seeds or majority albino can occur. A greater sample of seeds is more likely to achieve the expected ratio.

**5** Based on your individual and class results, what is the mode of inheritance for green pigmentation in barley?

Answer*:* Autosomal dominant.

**6** From a visual standpoint, homozygous green barley plants are indistinguishable from heterozygotes. To identify the genotype of an individual plant showing the dominant characteristic, a geneticist undertakes a test cross. Describe a test cross.

Answer*:* A test cross allows geneticists to determine the genotype of an organism, revealing all the alleles carried by the organism. To determine whether a plant exhibiting a dominant trait is homozygous or heterozygous for a specific allele, a geneticist can cross the plant with a plant of the same species that is homozygous for the recessive trait. The offspring of the test cross are then examined. If any of the offspring are recessive, the parent is identified as heterozygous for the allele. If only phenotypically dominant offspring are produced, then the parent is assumed homozygous dominant for the allele.

In the case of the barley, which self-pollinates asexually, the seeds are not crossed but instead planted at a distance from each other, so the growths are kept separate. Once they have grown, the plants from an individual seed are harvested in a bunch and kept separate from the other plants so that each set of seeds will be known to be from the one parent. From that harvest, several seeds are germinated until it is possible to distinguish green seedlings from white. If some are white, the parent is known to have been heterozygous; if none are white, the parent is assumed to have been homozygous dominant. It is impossible to grow homozygous recessive seeds beyond the germination stage.

If only phenotypically dominant offspring are present, some students may observe that we can still only say the parent is assumed to be homozygous, as statistical anomalies are possible in that they might be heterozygous but still only pass on the dominant allele.

**7** If presented with 120 seedlings, approximately how many would you expect to be green? Show your working out.

Answer*:* Student answers should state that approximately 90 seeds would be green. To come to this conclusion students should divide 120 by 4 to get 30 and then multiply that answer by 3.

Taking it further

**1** In this investigation you have conducted a monohybrid cross. What type of investigations could you conduct to demonstrate a dihybrid cross and a sex-linked cross?

Answer*:* To conduct a dihybrid cross from one parent who is homozygous dominant in both genes and the other who is homozygous recessive in both, e.g. AABB and aabb, the first generation would all be heterozygous in both genes, but showing the dominant phenotype. Cross the first generation together and the second generation should demonstrate the phenotypes in the ratio 9:3:3:1 (AB:Ab:aB:ab). The experiment could start with the first generation, i.e. both parents having both dominant phenotypes but with the genotype AaBb.

To conduct a sex-linked cross, e.g. in drosophila eyes (red vs white):

The white-eye gene is on the *X*-chromosome and is recessive. Cross a white-eyed female with a red-eyed male OR a red-eyed female with a white-eyed male OR run both in tandem.

Example 1:

White-eyed female (*xx*) and red-eyed male (*XY*)

First generation

|  |  |  |
| --- | --- | --- |
|  | *X* | *Y* |
| *x* | *xX* (red) | *xY* (white) |
| *x* | *xX* (red) | *xY* (white) |

All males have white eyes, all females red.

Second generation

|  |  |  |
| --- | --- | --- |
|  | *x* | *Y* |
| *x* | *xx* (white) | *xY* (white) |
| *X* | *Xx* (red) | *XY* (red) |

50% of each sex will have red eyes, 50% white

Example 2:

Red-eyed homozygous female (*XX*) and white-eyed male (*xY*)

First generation

|  |  |  |
| --- | --- | --- |
|  | *x* | *Y* |
| *X* | *Xx* (red) | *XY* (red) |
| *X* | *Xx* (red) | *XY* (white) |

All flies will have red eyes.

Second generation

|  |  |  |
| --- | --- | --- |
|  | *X* | *Y* |
| *X* | *XX* (red) | *XY* (red) |
| *x* | *xX* (red) | *xY* (white) |

All females will have red eyes, 50% of males will have red eyes, 50% white

**2** Calculate the chi-square value for this experiment. Do your observed frequencies deviate significantly from the expected frequency of this cross?

Answer*:* Collate all results.

Create a contingency table of the observed values (O), for example:

|  |  |  |  |
| --- | --- | --- | --- |
|  | **Green** | **White** | **Total** |
| Group A | 17 | 5 | 22 |
| Group B | 15 | 6 | 21 |
| Group C | 17 | 6 | 23 |
| Group D | 14 | 5 | 19 |
| Total | 63 | 22 | 85 |

Multiply each row total by each column total and divide by overall total to get the ‘expected’ values (E), e.g.:

|  |  |  |  |
| --- | --- | --- | --- |
|  | **Green** | **White** | **Total** |
| Group A | (63x22)/85 = **16.31** | (22x22)/85 = **5.69** | 22 |
| Group B | (63x21)/85 = **15.56** | (22x21)85 = **5.44** | 19 |
| Group C | (63x23)/85 = **17.05** | (22x23)/85 = **5.95** | 23 |
| Group D | (63x19)/85 = **14.08** | (22x19)/85 = **4.92** | 19 |
| Total | 63 | 22 | 85 |

Use the formula (O-E)2/E, then add up the resulting values, e.g.:

|  |  |  |  |
| --- | --- | --- | --- |
|  | **Green** | **White** | **Total** |
| Group A | (17-16.31)2/16.31 = **0.029** | (5-5.69)2/5.69 = **0.084** |  |
| Group B | (15-15.56)2/15.56 = **0.020** | (6-5.44)2/5.44= **0.058** |  |
| Group C | (17-17.05)2/17.05 = **0.000** | (6-5.95)2/5.95= **0.000** |  |
| Group D | (14-14.08)2/14.08 = **0.090** | (5-4.92)2/4.92 = **0.001** |  |
| Total |  |  | **0.282** |

The chi-square value is 0.282 in this example.

Student answers regarding their results versus expected results will vary.

Chapter 15 review questions

Recall

**1** Define the following terms:

**a** pure-breeding

Answer*:* Pure-breeding individuals are those that produce the same characteristic in each succeeding generation when bred among themselves. Pure-breeding individuals are homozygous – the two alleles for the gene under consideration are the same.

**b** progeny

Answer*:* Progeny are the offspring or children.

**c** hybrid

Answer*:* A hybrid is an individual who is heterozygous – the individual possesses two different alleles for the same gene.

**d** dominant

Answer*:* A dominant allele is one that masks the effect of another allele. The characteristic it produces is called the dominant characteristic and is seen in the phenotype.

**e** recessive

Answer*:* A recessive allele is one that is masked by another allele. The recessive characteristic is only seen in the phenotype when there is no dominant allele present.

**f** co-dominant

Answer*:* Co-dominance is when neither allele dominates (or masks the effects of) the alternative allele. When both alleles are present, the organism possesses either both characteristics or a blend of the two.

**g** carrier

Answer*:*  An individual who carries a recessive allele in their genotype that is not expressed in their phenotype.

**h** hemizygous

Answer*:*  Having no allelic counterpart, occurs with alleles in the *X* chromosome in males.

**i** first filial generation.

Answer*:* The first filial generation, or F1, are the offspring of the parental generation; that is, they are the first generation of offspring of a series of crosses.

**2** Briefly describe what is meant by the principle of segregation.

Answer*:* The principle of segregation states that the members of a pair of alleles separate (or segregate) during the formation of gametes.

**3** Describe the difference in appearance between the *X* and *Y* chromosomes.

Answer*:* The *X* chromosome is larger than the *Y* chromosome and contains more genes than the *Y* chromosome.

**4 a** What are autosomes?

Answer*:* Autosomes are the non-sex chromosomes – chromosome numbers 1–22.

**b** How many autosomes occur in:

**i** each normal human cell?

Answer*:* There are 44 autosomes in normal human cells

**ii** each sperm or egg?

Answer*:* There are 22 autosomes in each sperm and egg

**5 a** What are sex-linked characteristics?

Answer*:* Sex-linked characteristics are controlled by genes that are located on the *X* chromosome. They show different patterns of inheritance in the two sexes.

**b** Give examples of such characteristics.

Answer*:* Red-green colour blindness, haemophilia and Duchenne muscular dystrophy

**c** Why are sex-linked characteristics also called *X*-linked characteristics?

Answer: Sex-linked characteristics are found on the *X* chromosome. Their inheritance relies on the sex of the individual.

**6** List five rules that must be observed when constructing a pedigree.

Answer*:* Any five of:

a Use the symbols 🞏 for male and ⭘ for female.

b Indicate individuals with the trait with ◼ or ⚫.

c Represent a union between a male and a female by a horizontal line between the two individuals.



d Children are represented by a vertical line extending from the horizontal line joining the couple.



e The order in which children are born is shown with the eldest on the left and the youngest on the right.

f A union between two close relatives is shown by two horizontal lines.



**7** Describe the pattern of inheritance of the following disorders:

**a** cystic fibrosis

Answer*:* Cystic fibrosis is autosomal recessive.

**b** red–green colour blindness

Answer*:* Colour blindness is X-linked recessive.

**c** Huntington’s disease

Answer*:*  Huntington’s disease is autosomal dominant

**d** phenylketonuria.

Answer*:* Phenylketonuria is autosomal recessive.

**8** Phenylketonuria is one genetic disorder discussed in this chapter. Briefly outline:

**a** the symptoms caused

Answer*:* Phenylketonuria causes damage to the brain, intellectual disability, epileptic seizures and abnormal skin pigmentation.

**b** how it can be identified in newborn infants

Answer*:* Diagnosis in newborns is by a blood test and the blood is taken by a heel stick.

**c** the treatment that is given.

Answer*:* Treatment is a special diet that restricts the intake of the amino acid phenylalanine, replacing it with a substitute.

**9** **a** Describe what is meant by the term ‘DNA profile’.

Answer*:* To create a ‘DNA profile’ a sample of the DNA is cut at particular base sequences and placed on a bed of gel. Electrophoresis results in the pieces of DNA forming a banding pattern dependent on the size of the DNA fragment. This banded picture is the person’s DNA profile or fingerprint.

**b** List benefits that have arisen for those with hereditary disease, from the use of DNA profiling.

Answer*:* Benefits are:

* used to identify if people carry the gene for cystic fibrosis, Huntington’s disease, sickle cell anaemia, some cancers and other genetic diseases
* used to identify alleles that have mutated and may increase the risk of some conditions
* used for early identification of some hereditary diseases so that early treatment can occur.

**c** How has DNA profiling contributed to determination of parentage and ancestry?

Answer*:* DNA profiling has contributed to the determination of parentage and ancestry. By comparing DNA fingerprints, scientists are able to search for specific markers on the strands of DNA. They look for the number of repeats at each marker. If 10 different markers on different chromosomes are examined, there is only a 1 in a million chance that two people will have the same number of repeats, except for identical twins. The more markers that match, the more likely it is that the two individuals are related.

**d** State one ethical consideration with genetic profiling.

Answer*:* Any one of:

* Ownership: To whom does the genetic profile belong? Is it the individual, the laboratory that carried out the testing, medical authorities who wish to use the information, or to some other individual or group? The major issue that arises is the individual’s right to privacy.
* Best interest: Is it in an individual’s best interest to inform them of a genetic disease? Such information may lead to unnecessary anxiety, and possibly stigma, and it may change the person’s perception of wellness.
* Discrimination: There is the possibility that genetic information about an individual could be misused to their detriment. For example, health insurers could use genetic information and the potential risk of disease to adjust insurance premiums. On the other hand, an employer may prefer a particular genetic profile and rank applicants for a position accordingly.
* Potential for covert use: There have been cases where a person’s DNA has been obtained without their knowledge. This has occurred in cases where paternity has been questioned. In some countries, divorce has resulted after a father has chosen not to continue bringing up a child he may have thought was his before doing a DNA test.

Explain

**10** Using examples, distinguish between:

**a** homozygous and heterozygous

Answer*:* Homozygous (for example, TT) is when there are two identical alleles for a characteristic, while heterozygous (for example, Tt) is when there are two different alleles present for one characteristic.

**b** phenotype and genotype

Answer*:* The phenotype is the physical expression of a gene – the appearance of the organism – while genotype is the genetic make-up of the organism.

**c** allele and gene.

Answer*:* A gene is the factor that determines a specific characteristic. It is located on a chromosome.

Alleles are the alternative forms of the gene. There are often two alleles for a gene, but there may be more.

**11** Use an example to explain co-dominance.

Answer*:* Co-dominance is when neither allele dominates (or masks the effects of) the alternative allele.

When both alleles are present, the organism possesses either both characteristics or a blend of the two.

Example: When a homozygous red flower is crossed with a homozygous white flower, all resulting heterozygous offspring have pink flowers.

**12** Explain how the sex of a child is determined at the time of fertilisation.

Answer*:* The male gamete carries either an *X* sex chromosome or a *Y* sex chromosome. The type of sperm (*X* or *Y*) that fertilises the female gamete will determine the sex of the resulting child, as the female gamete can only carry an *X* sex chromosome.

|  |  |
| --- | --- |
| Gametes | Eggs |
| *X* | *X* |
| Sperm | *X* | *XX* | *XX* |
| *Y* | *XY* | *XY* |

**13** Explain how multiple alleles are important in blood groups.

Answer*:* Multiple alleles occurs when more than two alleles for a gene are located at the same position on a chromosome. In blood typing this has the allele for A, the allele for B and the allele for o all occurring at the same position. A person can inherit any two of the three alleles, and this presents more than two phenotypic representations for blood types.

**14** Explain why a father with an X-linked condition is not able to pass the characteristic to his sons.

Answer*:* A father with an X-linked condition cannot pass it on to his sons, because his male children must inherit his *Y* chromosome, and therefore cannot inherit his *X* chromosome.

15 Describe what is meant by ‘genetic counselling’, and discuss how it may assist people in deciding whether to have a child or to continue with a pregnancy.

Answer*:* Genetic counselling is advice given to a person or couple in relation to the risk that an inherited condition may affect their offspring. Usually genetic counselling is sought if there is a history of an inherited condition in the family of one or both of the prospective parents. The genetic counsellor can often advise the couple of the probability of an inherited condition occurring in their children. With this knowledge the couple can then decide whether they wish to have children or to continue with a pregnancy.

Apply

**16** Using your own family as an example, explain the difference between a first filial generation and a second filial generation.

Answer*:* The first filial generation is the offspring produced by my parents. The second filial generation will be the offspring produced by me or my siblings.

Alternatively, parents represent the first filial generation from the grandparents, and the children are the second filial generation. Student responses will vary according to their family.

**17 a** What is probability?

Answer*:* Probability is the chance that a particular event will occur.

**b** If two people with normal skincolouring, each with the recessiveallele for albinism, have a child, whatis the probability of the child being analbino?

Answer*:*  There is a 25% chance the child will be an albino.

**18** Why do couples who are first cousins have a slightly higher risk of having a child with an inherited disorder than unrelated couples?

Answer*:* Related couples have an increased chance of passing on an inherited disorder that they may both be carriers for, because both may have received the allele in question from a common ancestor.

**19** In garden peas, round seed shape is dominant to wrinkled seed shape. Pure-breeding round seed plants were crossed with pure-breeding wrinkled seed plants. Determine the expected genotypes and phenotypes of the F1 and F2, and the expected proportions.

Answer*:* *R* = round seed; *r* = wrinkled seed

|  |  |  |
| --- | --- | --- |
| *RR* | × | *rr* |
|  | *R* | *R* |
| *r* | *Rr* | *Rr* |
| *r* | *Rr* | *Rr* |

F1 generation: genotypes: 100% *Rr*

F1 phenotypes: 100% round seeds

|  |  |  |
| --- | --- | --- |
| *Rr* | × | *Rr* |
|  | *R* | *r* |
| *R* | *RR* | *Rr* |
| *r* | *Rr* | *rr* |

F2 generation: genotypes: 25% *RR*, 50% *Rr* and 25% *rr*

F2 phenotypes: 75% round seeds and 25% wrinkled seeds

**20** In humans, normal melanin production is dominant to albino, which produces white hair and pink eyes. The first child born to a married couple with normal pigmentation is an albino. Calculate the probability that the second child will also be an albino. Give a clear explanation for your results.

Answer*:* If both parents have normal pigmentation, but produce a child with albino pigmentation, then they must both be carriers, thus heterozygous (*Aa*, where *A* = normal pigmentation, and *a* = albino).

Because each birth is an independent event, the probability that the couple will have an albino child is not influenced by the pigmentation of any previous children. It therefore follows that there is a 25% chance that the second child will be albino (*aa*).

|  |  |  |
| --- | --- | --- |
| *Aa* | × | *Aa* |
|  | *A* | *a* |
| *A* | *AA* | *Aa* |
| *a* | *Aa* | *aa* |

21 In guinea pigs, black fur colour is dominant over white fur colour. How could an animal breeder test whether a black guinea pig is homozygous or heterozygous?

Answer*:* The breeder could cross a black guinea pig with a white guinea pig. A white guinea pig’s genotype is known from its phenotype – it must be homozygous recessive (homozygous for the white allele). If there are white offspring, then the black guinea pig is heterozygous. If there were no white offspring, it is more likely that the black guinea pig is homozygous.

**22** In humans, free earlobes are dominant over attached earlobes. A woman heterozygous for free earlobes marries a man with attached earlobes. Use a Punnett square to determine their chance of producing children with attached earlobes.

Answer*:* *F* = free ear lobes; *f* = attached ear lobes

|  |  |  |
| --- | --- | --- |
| *Ff* | × | *Ff* |
|  | *F* | *f* |
| *f* | *Ff* | *ff* |
| *f* | *Ff* | *ff* |

There is a 50% chance of children with attached earlobes.

23 In many families, a Roman-shaped nose is dominant to a straight nose. If a man from a family pure-breeding for a Roman nose has children with a woman from a family pure-breeding for a straight nose, what would they look like? If one of the children has children with a person from a family with a long history of straight noses, what types of noses would you expect the grandchildren to possess and in what proportions?

Answer*:* *R* = Roman-shaped nose; *r* = straight-shaped nose

A man pure-breeding for Roman nose and a woman pure-breeding for straight nose:

|  |  |  |
| --- | --- | --- |
| *RR* | × | *rr* |
|  | *R* | *R* |
| *r* | *Rr* | *Rr* |
| *r* | *Rr* | *Rr* |

All the children would have Roman-shaped noses.

A child of above couple and a person pure-breeding for straight nose:

|  |  |  |
| --- | --- | --- |
| *Rr* | × | *rr* |
|  | *R* | *r* |
| *r* | *Rr* | *rr* |
| *r* | *Rr* | *rr* |

Genotypes: 50% *Rr* and 50% *rr*

Phenotypes: 50% straight nose and 50% Roman nose

24 When plants that are pure-breeding for wrinkled seeds are crossed with plants that are pure-breeding for round seeds, the ratio of genotypes in the F2 is 1:2:1 and the ratio of phenotypes is 3:1. Explain what causes the genotypic ratio to differ from the phenotypic ratio.

Answer*:* The phenotypic and genotypic ratios differ because seeds with round seed shape are a combination of heterozygotes (*Rr*) and homozygotes (*RR*). The dominant allele masks the effect of the recessive allele, so both homozygous dominant and heterozygous genotypes have the round phenotype.

F2 generation:

|  |  |  |
| --- | --- | --- |
| *Rr* | × | *Rr* |
|  | *R* | *r* |
| *R* | *RR* | *Rr* |
| *r* | *Rr* | *rr* |

Genotypes: 25% *RR*, 50% *Rr* and 25% *rr*

Phenotypes: 75% round seeds and 25% wrinkled seeds

**25** If a human male with blood group M has children with a female with blood group N, what blood groups would they possess? If one of the children has children with a person with blood group M, what blood groups could the grandchildren possess? Construct the crosses for each of these matings. List the genotypes and phenotypes that would be expected, and the probability of obtaining each genotype and phenotype.

Answer*:* First mating: *BMBM* × *BNBN*:

|  |  |  |
| --- | --- | --- |
|  | *BM* | *BM* |
| *BN* | *BMBN* | *BMBN* |
| *BN* | *BMBN* | *BMBN* |

Probabilities:

• Genotypes of children: 100% *BMBN*

• Phenotypes of children: 100% type MN blood group

Second mating: *BMBN* × *BMBM*:

|  |  |  |
| --- | --- | --- |
|  | *BM* | *BN* |
| *BM* | *BMBM* | *BMBN* |
| *BM* | *BMBM* | *BMBN* |

Probabilities:

• Genotypes of grandchildren: 50% *BMBN* and 50% *BMBM*

• Phenotypes of grandchildren: 50% type MN blood group and 50% type M blood group

**26** A woman from a family with no history of haemophilia marries a man who is a haemophiliac. What is the probability that they will produce:

Answer*:* Possible genotypes for a mating between *XHXH*× *XhY*, where:

*H* = no haemophilia

*h* = haemophilia

|  |  |  |
| --- | --- | --- |
|  | *XH* | *XH* |
| *Xh* | *XHXh* | *XHXh* |
| *Y* | *XHY* | *XHY* |

**a** sons with normal blood clotting?

Answer*:* 100% – all sons will have normal blood clotting.

**b** sons with haemophilia?

Answer*:* 0% – no sons will have haemophilia.

**c** daughters who are carriers of haemophilia?

Answer*:* 100% – all daughters will be carriers.

**d** daughters who will be haemophiliacs?

Answer*:* 0% – no daughters will be haemophiliacs.

**27** Red–green colour blindness is a sex-linked characteristic. Under what circumstances would a couple produce daughters who all had normal vision and sons who were all colour blind? Describe the genotypes of both parents and all the children.

Answer*:* The circumstances under which a couple would produce daughters who all had normal vision and sons who would all be colour blind would occur when a male with normal vision produced children with a colour blind female, where *b* = colour blind; *B* = normal vision.

Possible genotypes for a mating between *XbXb* × *XBY*:

|  |  |  |
| --- | --- | --- |
|  | *Xb* | *Xb* |
| *XB* | *XBXb* | *XBXb* |
| *Y* | *XbY* | *XbY* |

Parents:

• Genotypes: *XbXb* and *XBY*

• Phenotypes: The mother is colour blind and the father has normal vision.

Children:

• Genotypes: males will be *XbY*; females will be *XBXb*;

• Phenotypes: colour blind boys and normal vision girls.

**28** The first child born to a married couple with normal vision is a male with red– green colour blindness. Calculate the probability that their second child will also be colour blind. Give a clear explanation for your answer. Remember that red–green colour blindness is *X*-linked.

Answer*:* Because the first child was colour blind, he must have received the affected allele from his mother, so his mother must be a carrier for the colour blindness allele.

Where *B* = normal vision; *b* = colour blind.

|  |  |  |
| --- | --- | --- |
|  |  | Female gametes |
|  |  | *XB* | *Xb* |
| Male gametes | *XB* | *XB XB* | *XB Xb* |
| *Y* | *XBY* | *XbY* |

Genotypes of offspring: 25% *XBXB*, 25% *XBXb*, 25% *XBY* and 25% *XbY*

Phenotypes of offspring: All females will have normal vision and there is a 50% probability that a male child will be colour blind.

There is a 25% chance the second child will be a male and will be colour blind. The genotype of the first birth has no bearing on the second, because the fertilisation of gametes to form each child is an independent event.

**29** In the United States, about 6 in every 100 children whose parents are first cousins die before the age of 10 years. Where the parents are unrelated, the figure is about 2.5 in every 100. Can you suggest reasons for this big difference in mortality for the first 10 years of life?

Answer*:* If the parents are related, there is a higher chance that the children will have inherited two defective alleles for a condition than if the parents are not related. The chances of cousins both being carriers for a recessive, disease-causing allele is greater than for unrelated people because cousins share common ancestors.

**30 a** Chloe is Rh+ but her brother Jason is Rh-. Both Chloe’s parents are Rh+. What is the probability that Chloe is a carrier for the recessive Rh allele?

Answer*:* Both of Chloe’s parents are heterozygous. We know this because they are both Rhesus-positive, but their son, Jason, is Rhesus-negative.

*R* = Rhesus-positive

*r* = Rhesus-negative

|  |  |  |  |
| --- | --- | --- | --- |
| Parents: | *Rr* | × | *Rr* |
|  |  | *R* | *r* |
|  | *R* | *RR* | *Rr* |
|  | *r* | *Rr* | *rr* |

We already know that Chloe is Rhesus-positive so we can exclude the Rhesus-negative possibility. Thus there is a two-thirds chance that Chloe is a carrier.

**b** Chloe married Mitchell and they had a daughter, Zara. What is the probability that Zara is Rh- if:

**i** Chloe is a carrier for the recessive allele?

Answer*:* This depends on Mitchell’s genotype.

Possibility 1: Mitchell *RR* × Chloe *Rr*. There is 0% chance that Zara is Rhesus-negative:

|  |  |  |
| --- | --- | --- |
|  | *R* | *r* |
| *R* | *RR* | *Rr* |
| *R* | *RR* | *Rr* |

Possibility 2: Mitchell *Rr* × Chloe *Rr*. There is a 25% chance that Zara is Rhesus-negative:

|  |  |  |
| --- | --- | --- |
|  | *R* | *r* |
| *R* | *RR* | *Rr* |
| *R* | *Rr* | *rr* |

Possibility 3: Mitchell *rr* × Chloe *Rr*. There is a 50% chance that Zara is Rhesus-negative:

|  |  |  |
| --- | --- | --- |
|  | *R* | *r* |
| *r* | *Rr* | *rr* |
| *r* | *Rr* | *rr* |

**ii** Chloe is not a carrier for the recessive allele?

Answer*:* To be Rhesus-negative, Zara needs two copies of the Rh– allele, one from each parent. If Chloe does not carry the recessive allele, then Zara cannot be Rhesus-negative. The possibility is 0% regardless of what Mitchell’s genotype is.

**31** Examine the pedigree on the next page of families where some individuals have the sickle-cell allele.

**a** Which individuals have sickle-cell anaemia?

Answer*:* The individuals with sickle-cell anaemia are I 3, II 3, and II 7.

**b** Which individuals must have sickle-cell trait?

Answer*:* The individuals who must have the sickle-cell trait are I 1, I 2, I 4 and II 6.

**c** Couple II 3 and II 4 are thinking of having a child. If you were a genetic counsellor, what advice would you give them regarding the possibility of having a child with sickle-cell anaemia or sickle-cell trait?

Answer*:* The individual II 3 has sickle-cell anaemia, which means she is homozygous and will therefore pass on the affected allele to her children. If individual II 4 was carrying the sickle-cell trait, then there would be a 50% chance of their children having the sickle-cell trait and a 50% chance of them having sickle-cell anaemia. If II 4 was not a carrier and therefore homozygous for normal red blood cells, then their children would all be carriers of the trait.

d None of the children of II 5 and II 6 have sickle-cell anaemia. Does this mean that the allele for sickle-cell anaemia no longer occurs in that branch of the family?

Answer*:* Sickle-cell anaemia could still occur in further generations of the family of II 5 and II 6 because II 6 has the sickle-cell trait and he may have passed the affected allele on to some or all of his children.

**32** In October 2013, Irish police seized two children from Roma (Gypsy) families, claiming that they were too blonde to be the offspring of their dark-haired parents. DNA testing proved that the dark-haired couples were indeed their parents.

**a** How is it possible that two people with dark hair could have children with blonde hair?

Answer*:* Two people with dark hair could produce children with blonde hair if each parent carried the recessive allele for blonde hair, and the child inherited a recessive allele from each parent.

**b** What is the probability that one of the couples could have another child with blonde hair?

Answer*:* The probability that one of the couples could have another child with blonde hair is 25%, 0.25 or 1/4.

*D* = dark hair; *d* = blonde hair

|  |  |  |
| --- | --- | --- |
|  | *D* | *d* |
| *D* | *DD* | *Dd* |
| *d* | *Dd* | *dd* |

Extend

**33** People with Huntington’s disease often have children, even though their children will have at least a 50% chance of inheriting the disease. With such a high probability of passing the disease on, why do such people continue to have children?

Answer*:* Huntington’s disease does not become apparent until much later in life, and the people concerned have usually already had children by then.

**34** With DNA profiling, genetically inherited diseases can be detected at an early age. Discuss the advantages of the early detection of a particular genetic disease.

Answer*:* Generally, early detection allows preventative therapies, strengthening of the individual, medical procedures, reduction in symptoms and management of the disease. The aim is to have early detection and eventually a cure before symptoms appear. Early detection may also help couples to decide whether to terminate a pregnancy.

**35** Describe why it is impossible for parents who have the blood groups A and AB to produce children with blood group O.

Answer*:* It is impossible for parents with blood groups A and AB to produce children with blood group O because the parent with blood group A can only pass on an *IA* or *i* allele; while the parent with blood group AB can only pass on an *IA* or *IB* allele. For a child to have blood group O they must receive the *i* allele from both their parents, and this is not possible in this situation.

**36** Charlie Chaplin, a famous comedian of the silent screen, was taken to court in 1944 by a young starlet, Joan Barry. She claimed that Chaplin was the father of her child, and the court ruled in her favour. Blood group data was not admissible evidence at the time of that trial. However, if you were the judge, how would you have decided? The baby was blood group B, the mother A and Chaplin O. Give genetic reasons for your decision.

Answer*:* If I had been the judge, I would have ruled that the child could not have been fathered by Chaplin. The baby would have had the genotype of either *IBIB* or *IBi*; while the mother could have had genotypes *IAIA* or *IAi*. Therefore, for the baby to be blood group B, its father would have had to pass on at least one *IB* allele. Chaplin could not have done this because his only possible genotype was *ii*.

**37** A woman has a brother with Duchenne muscular dystrophy. What information could be given to the woman about the risk of her having a child with Duchenne muscular dystrophy?

Answer*:* If the woman has a brother with the disease there is a 50% chance that she has inherited the defective allele. If she is a carrier, any male children will then have a 50% chance of inheriting the defective allele and suffering from the disease. The woman should receive genetic counselling to assist her in deciding if she wants to have children, and to know all the demands of having a child with Duchenne muscular dystrophy, who will eventually die prematurely. The counsellor may suggest undergoing genetic testing for the condition.

**38** In what situations would you be able to deduce a person’s genotype by determining their phenotype? Give examples using ABO blood groups.

Answer*:* It is possible to deduce a person’s genotype from their phenotype when the observable characteristic is a result of co-dominance or recessive inheritance. For example:

• AB blood group (phenotype) must result from *IAIB* (genotype)

• O blood group must result from *ii*.

**39** DNA profiling raises ethical questions. Discuss the ethical issues of the following situations.

**a** A wealthy grandparent suspects that a grandchild is not genetically related to her and plans to disinherit him if that is the case.

**b** A devoted fan is willing to pay a high price to purchase the genetic information of their favourite celebrity.

**c** A political party is interested in discovering and publicising any predispositions to disease that might render a candidate of the opposing party unsuitable for election.

Answer*:* Independent research, so answers will vary. However, the second two situations are highly unethical, and the first, highly questionable depending on how the grandparent intends to gather the information required to make her decision.