|  |  |  |  |
| --- | --- | --- | --- |
| The Hh blood group system is controlled by one gene locus with two alleles.The homozygous recessive genotype produces the Bombay phenotype, resulting in a very rare blood group, in which no antigen is expressed.The Bombay phenotype is very rare. One person in 250 000 of the world's population is estimated to have the Bombay phenotype.1. Using the Hardy-Weinberg equations, calculate the **percentage** of the world's population who carry one copy of the recessive allele.

|  |  |  |
| --- | --- | --- |
| *p* + *q* = 1 |  | *p*2 + 2*pq* + *q*2 = 1 |

1. Show each step in your working. Give your answer to **one significant figure**.
2. percentage \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ \_ **[4]**

The Bombay phenotype is more common in some regions of India, where it can occur in one in 10000 people. Researchers have suggested that the Bombay phenotype is more common in these regions because of the practice of endogamy, in which marriage occurs only between people within the same tribe or small social group.Suggest why endogamy has increased the frequency of the Bombay phenotype.    **[2]** |
|  |

**2.** In the flour beetle, the allele for red body colour (**R**) is dominant to the allele for black body colour (**r**). A mixed culture of red beetles and black beetles was kept in a container in the laboratory under optimal breeding conditions. After one year, there were 149 red beetles and 84 black beetles in the container.

**1**

(a) Use the Hardy-Weinberg equation to calculate the expected percentage of heterozygous red beetles in this population.

Answer: .............................................. (3)

(b) Several assumptions are made when using the Hardy-Weinberg equation. Give **two** of these.

1..................................................................................................................................

2.................................................................................................................................. (2)

(Total 5 marks)

**3**. A recessive allele gives increased resistance to infection by the malarial parasite. In a population, the proportion of babies born who are homozygous for this allele is 0.01.

Use the Hardy-Weinberg equation to calculate the expected proportion of heterozygotes in this population.

**4**. Huntington’s disease is an inherited condition resulting in gradual degeneration of nerve cells in the brain. It is caused by a dominant allele but there are usually no symptoms until the person is at least 30 years old. It is very rare in most populations but in one isolated area of South America, 48% of the population possess a genotype which gives rise to Huntington’s disease. Many of the inhabitants of this area can trace their origins back to a common ancestor 200 years ago.

(a) Use the Hardy-Weinberg equation to estimate the percentage of this population which is heterozygous for Huntington’s disease. Show your working.

(b) Outline why Huntington’s disease has not been eliminated from this population by natural selection.

**5.** (a) Explain what is meant by

a *recessive* allele;

....................................................................................................................................

....................................................................................................................................

a *sex-linked* gene.

....................................................................................................................................

.................................................................................................................................... (2)

(b) Nail-patella syndrome is an inherited condition caused by a single gene. Sufferers have abnormal nail growth and underdeveloped kneecaps. The pedigree shows how members of one family were affected by the syndrome.



Explain **one** piece of evidence from the pedigree which indicates that

(i) the allele for the nail-patella syndrome is dominant;

............................................................................................................................

............................................................................................................................

............................................................................................................................

............................................................................................................................

(2)

(ii) the gene is **not** sex-linked.

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............................................................................................................................

(2)

(Total 6 marks)

**6.** The inheritance of the ABO blood groups is an example of multiple allele inheritance and is controlled by three alleles of a single gene, **IO, IA** and **IB**

 These three alleles determine the activity of an enzyme which modifies the structure of an antigen on the cell surface membrane of the red blood cells. This is summarised below.

****

 (a) The allele **IO** is recessive to both alleles **IA** and **IB**. Alleles **IA** and **IB** are codominant, that is, they are both expressed in the phenotype.

The diagram shows the inheritance of ABO blood groups in one family.



(i) Give the blood group genotype or genotypes of the sperm cells produced by individual 2.

..........................................................................................................................

 (1)

(ii) Give the blood group phenotype of individual 1.

..........................................................................................................................

 (1)

(iii) Calculate the probability that the next child produced by individuals 4and 5will be a boy with blood group **A**. Show your working.

..........................................................................................................................

 ..........................................................................................................................

 (2)

 (Total 4 marks)

**7.** The allele for Rhesus positive, **R**, is dominant to that for Rhesus negative, **r**.
Haemophilia is a sex-linked condition. The allele for haemophilia, **h**, is recessive to the allele for normal blood clotting, **H**, and is carried on the X–chromosome.
The diagram shows the Rhesus blood group phenotypes in a family tree where some individuals have haemophilia.



(a) (i) Use the information in the diagram to give **one** piece of evidence that the allele for the Rhesus negative condition is recessive.

..........................................................................................................................

.......................................................................................................................... (1)

(ii) Explain the evidence from the cross between individuals 3 and 4 that the gene controlling Rhesus blood group is **not** sex-linked.

..........................................................................................................................

.......................................................................................................................... (2)

(b) Give the full genotype of

(i) individual 6;

.......................................................................................................................... (1)

(ii) individual 12.

.......................................................................................................................... (1)

(c) What is the probability that the next child of couple 10 and 11 will have the same genotype as the first child? Show your working.

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..................................................................................................................................... (3)

(Total 8 marks)

**8.**          In fruit flies, the allele for grey body, **G**, is dominant to the allele for ebony body, **g**, and the allele for normal wings, **N**, is dominant to the allele for vestigial wings, **n**. Vestigial-winged flies, heterozygous for grey body colour, were crossed with ebony-bodied flies, heterozygous for normal wings.

          Complete the genetic diagram to show the genotypes and phenotypes in this cross.

          *Parental phenotypes* Grey body, vestigial wings          Ebony body, normal wings

*Parental genotypes* ..............................                        ...............................

*Gamete genotypes* ..............................                        ...............................

*Offspring genotypes* ..............................................................................................

*Offspring phenotypes* ............................................................................................

**(Total 4 marks)**

**9.**          In a breed of cattle the **H** allele for the hornless condition is dominant to the **h** allele for the horned condition. In the same breed of cattle the two alleles **CR** (red) and **CW** (white) control coat colour. When red cattle were crossed with white cattle all the offspring were roan. Roan cattle have a mixture of red and white hairs.

(a)     Explain what is meant by a *dominant* allele.

......................................................................................................................

...................................................................................................................... **(1)**

(b)     Name the relationship between the two alleles that control coat colour.

...................................................................................................................... **(1)**

(c)     Horned, roan cattle were crossed with white cattle heterozygous for the hornless condition. Compete the genetic diagram to show the ratio of offspring phenotypes you would expect.

*Parental phenotypes*             Horned, roan               ×               hornless, white

*Parental genotypes*

*Gametes*

*Offspring genotypes*

*Offspring phenotypes*

*Ratio of offspring*

*phenotypes*

**(4)**

(d)     The semen of prize dairy bulls may be collected for in vitro fertilisation. The sperms in the semen can be separated so that all the calves produced are of the same sex. The two kinds of sperms differ by about 3% in DNA content.

(i)      Explain what causes the sperms of one kind to have 3% more DNA than sperms of the other kind.

.............................................................................................................

.............................................................................................................

.............................................................................................................

............................................................................................................. **(2)**

(ii)     Suggest **one** reason why farmers would want the calves to be all of the same sex.

.............................................................................................................

............................................................................................................. **(1)**

**(Total 9 marks)**

 **10.**          A sex-linked gene controls fur colour in cats. Ginger-coloured fur is controlled by the allele **G**, and black-coloured fur is controlled by the allele **g**. Some female cats have ginger and black patches of fur. They are described as tortoiseshell. Male cats cannot be tortoiseshell.

(a)     What is meant by a *sex-linked* gene?

......................................................................................................................

...................................................................................................................... **(1)**

(b)     A male cat with the genotype **Xg Y** mates with a tortoiseshell female.

(i)      Give the phenotype of the male.

............................................................................................................. **(1)**

(ii)     Give the genotype of the tortoiseshell female.

............................................................................................................. **(1)**

(iii)     Complete the genetic diagram to show the genotypes and the ratio of phenotypes expected in the offspring of this cross.

*Parents* Male                     Tortoiseshell female

*Parental genotypes* **Xg Y** ........................

P*arental gametes*

*Offspring genotypes*

*Offspring phenotypes*

*Ratio*  **(3)**

(c)     The effect of the **G** and **g** alleles is modified by another gene. This gene is not sex-linked and it has two alleles. The allele **d** changes the ginger colour to cream and the black colour to grey. The dominant allele **D** does not modify the effect of **G** or **g**.

A cream-coloured male cat mated with a black female whose genotype was **XgXg Dd**. Male kittens of two different colours were produced. Complete the genetic diagram.

*Parental* Cream-coloured                     Black
*phenotypes* male                              female

*Parental* .....................                        **XgXg Dd***genotypes*

          *Parental
gametes*

          *Male kitten
genotypes*

          *Male kitten
colours*

 **(3)**

**(Total 9 marks)**

**11.**          Coat colour in Labrador dogs is controlled by two different genes. Each gene has a dominant and a recessive allele. The two genes are inherited independently but the effects of the alleles interact to produce three different coat colours. The table gives four genotypes and the phenotypes they produce.

|  |  |
| --- | --- |
| **Genotype** | **Phenotype** |
| **BbEe** | black |
| **bbEe** | chocolate |
| **Bbee** | yellow |
| **bbee** | yellow |

(a)     What colour coat would you expect each of the following genotypes to give?

(i)      **BBEe** …………………………

(ii)     **bbEE** …………………………

**(2)**

(b)     A **BbEe** male was crossed with a **bbee** female. Complete the genetic diagram to show the ratio of offspring you would expect.

*Parental phenotypes*                      Black male           ×            Yellow female

*Parental genotypes*                             **BbEe**                                    **bbee**

*Gametes*

*Offspring genotypes*

*Offspring phenotypes*

*Ratio of offspring*

*phenotypes*

**(3)**

(c)     The yellow coat colour of Labrador dogs is due to the presence of the pigment phaeomelanin in the hairs. The black and chocolate coat colours are due to different amounts of another pigment, eumelanin, deposited in these hairs. The more eumelanin there is, the darker the hair. The diagram shows the action of genes **E** and **B** in producing the different coat colours.



Use this information to explain how

(i)      the genotype **bbee** produces a yellow coat colour;

.............................................................................................................

.............................................................................................................

............................................................................................................. **(2)**

(ii)     the genotype **BbEe** produces a black coat colour.

.............................................................................................................

.............................................................................................................

............................................................................................................. **(2)(Total 9 marks)**

**12**. Colour blindness is controlled by a gene on the X chromosome. The allele for colour blindness, **X**b, is recessive to the allele for normal colour vision, **X**B. The gene controlling the presence of a white streak in the hair is not sex linked, with the allele for the presence of a white streak, **H**, being dominant to the allele for the absence of a white streak, **h**.

(a) Explain why colour blindness is more common in men than in women.

....................................................................................................................................

....................................................................................................................................

....................................................................................................................................

.................................................................................................................................... (2)

(b) The diagram shows a family tree in which some of the individuals have colour blindness or have a white streak present in the hair.



(i) What are the genotypes of individuals **5** and **6**?

 Individual **5** .....................................................................................................

 Individual **6** ..................................................................................................... (2)

(ii) Give the possible genotypes of the gametes produced by

 individual **5**; .....................................................................................................

 individual **6**. ..................................................................................................... (1)

(iii) What is the probability that the first child of individuals **5** and **6** will be a colour blind boy with a white streak in his hair? Show your working.

Answer ............................................(2)

(Total 7 marks)

MARK SCHEME

**1.** **Answer:** i) q = √ 0.000004 = 0.002 p = 1 − 0.002 = 0.998

2pq = 2 × 0.998 × 0.002 = 0.003992 % to 1 significant figure = 0.003992 × 100 = 0.4 %

ii) **Any 2 from:**
small / decreased, gene pool
inbreeding
genetic drift
population / genetic, bottleneck

ii) **ALLOW** decreased genetic variation
**IGNORE** interbreeding
**IGNORE** references to (increased) homozygous recessive genotypes because this is implied by information provided earlier in the question

**2.** (a) EITHER: q2 = 0.36 / q = 0.6/0.61;
 p = (1 – 0.6) = 0.4/0.39;
 48/47.9 3 marks

(b) No selection;
random mating/no sexual selection;
large population/gene pool;
no emigration/immigration/no migration/isolated population;
no mutation;
equally viable gametes/all equally fertile;
generations do not overlap; max. 2 [5]

3. correct answer = 0.18;

p + q = 1 and p2 + 2pq + q2 = 1; 0.01 = q2 ; q = 0.1; p = 0.9

frequency of heterozygotes = 2pq = 2 x 0.1 x 0.9;

**4**. (a) (q2 = 0.52 / q = 0.72)

(p = 1 – 0.72 = 0.28)

p + q = 1 / p2 + 2pq + q2 = 1 ;

Answer = 2pq / use of appropriate numbers;

Answer = 40%;

(b) Genetic isolation / small gene pool / no immigration /no migration / in-breeding;

High probability of mating with person having H-allele;

Reproduction occurs before symptoms of disease areapparent;

Genetic argument – Hh x hh \_ 50% / Hh x Hh \_ 75% affectedoffspring;

No survival / selective disadvantage;

**5.** (a) recessive only expressed (in the phenotype) when homozygous 1

 sex-linked gene is on the X chromosome (in humans)/Y chromosome. 1

(b) (i) 3 and 4 produce unaffected male/8 / female/10, so mustcarry recessive;
but both affected by nail-patella which must be dominant. 2

(ii) 3 inherits X from mother, who is not affected;
if sex-linked, 3 would have nail-patella on X chr and would pass on to all female offspring;
10 is recessive female, so gene not sex-linked. max 2 [6]

**6.** (a) (i) **IA** and **IO**; 1

(ii) Blood group B; 1

(iii) Probability of child being boy indicated as 0.5and probability
of child being blood group A indicated as 0.5;
0.25or other correct expression; 2 [4]

**7.** (a) (i) specific cross identified - 3 ,4 & 8 or 10, 11 & 12 /Rh negative
phenotype produced from parents which are both Rh positive; 1

(ii) with sex linkage daughter cannot have (recessive) condition unless
male parent has the condition;
as male passes X chromosome to his daughter; 2

(b) (i) Rr XHXh;

(ii) rr XhY; 2

(c) P(rr) = ¼;P(XhY) = ¼;probability = 1/16/6.25%/0.0625;

OR

 Punnett square with first two marking points-
genotypes of gametes of one parent correct;
genotypes of gametes of other parent correct;
probability = 1/16/6.25%/0.0625; 3 [8]

 **8.**          Parental genotypes:               Gg nn                    gg Nn       ;
Gamete genotypes                 Gn      gn               gN            gn       ;

|  |  |  |
| --- | --- | --- |
|   | gN | gn |
|  Gn  |  Gg NnGrey, normal |  Gg nnGrey, vestigial |
|  gn  |  gg NnEbony, normal |  gg nnEbony, vestigial |

All offspring genotypes correct;

All offspring genotypes correctly derived; **[4]**

**9.**          (a)     is always expressed(in the phenotype) / produces (functional) proteins; **1**

(b)     codominance; **1**

(c)     *Parental geneotypes* -    hhCRCw,                       HhCwCw;

Gametes-                      

*Offspring geneotypes -* HhCRCw,   hhCRCw,  HhCwCw,   hhCwCw;

*Offspring pheneotypes -* hornless   horned   hornless   horned

          roan         roan      white       white
*Ratio of offspring* -          1               1            1               1; **4**

(d)     (i)      sperm(with more DNA) have X chromosome;
X is larger / has more genes than Y;

**2**

(ii)     female for milk / males for meat / male or female for breeding;

**1**

**[9]**

 **10.**          (a)     gene located on X / Y/ one sex chromosome;

*(allow gene on X or Y chromosome, not X and Y)*

**1**

(b)     (i)      black;

**1**

(ii)     **XGXg**;

*(lose this mark if the wrong genotype is given for the female in (iii))
(must show X chromosomes to gain the mark)*

**1**

correct parent gametes
(**Xg** and **Y** from male, **XG** and **Xg** from female);
correct offspring genotypes (**XgXg**, **XGXg**, **XGY**, **XgY**);
correct link of offspring genotypes with phenotypes;
**XgXg** black female
**XGXg** tortoiseshell female
**XGY** ginger male **XgY** black male

*(correct gametes, offspring genotypes and link with phenotypes based on incorrect**parent genotype = 3 marks)*

**3**

(c)     **XGY dd**; correct male kitten genotypes (**XgY Dd** and **XgY dd**);
correct link of kitten genotypes with phenotypes;

*(ignore female kittens)*

**XgY Dd**         black
**XgY dd**          grey

*(correct kitten genotypes and phenotypes based on incorrect parent genotype = 2 marks)*

**3**

**[9]**

**11.**          (a)     (i)      black;

**1**

(ii) chocolate;

**1**

(b)     **BE**, **Be**, **bE**, **be** and **be**;

**BbEe, Bbee, bbee, bbEe;**

1 black: 2 yellow: 1 chocolate;

**3**

(c)     (i)      no enzyme coded for when no dominant / **E** allele;

phaeomelanin not converted – (remains yellow);

**2**

(ii)     **E** allele results in enzyme producing eumelanin;

**B** allele - more eumelanin deposited in hairs;

**2**

**[9]**

**12**. (a) males are XY and females XX / males have one X chromosome and
females two X chromosomes;
males only have one allele (of the gene) present / recessive allele
always expressed;
colour blindness is masked in heterozygote / female needs 2 recessive
alleles to be colour blind; 2 max

(b) (i) 5 - hh Xb Y;
6 - Hh XB Xb ; 2

(ii) h Xb , h Y, and H XB, h XB, H Xb , hXb; 1

(iii) 1/8 or 12.5% or 0.125;;

 *either*

 genetic diagram to show genotypes Hh Xb Xb , Hh XBY, hh XB Xb,
hh XBY, HHXbXb, Hh XbY, hh Xb Xb; hh XbY;
1/8;
*or*P (boy) = 0.5, P (colour blind) = 0.5, P (white streak) = 0.5;
(0.5 × 0.5 × 0.5 =) 0.125; 2

[7]