Model Answers: Hard

1a

a) The number of offspring that would have a grey body colour and vestigial wings would be as follows:

- None / 0; [1 mark]

Explanation:

- Each offspring would receive at least one allele for long wings (**L**) from the parent that is homozygous for long wings (**LL**); [1 mark]
- The presence of the dominant / **L** / allele for long wings would mask the presence of the recessive / **l** / allele for vestigial wings (in the phenotype); [1 mark]

[Total: 3 marks]

The genotype of the two fruit flies would be:

Parent 1: GGLL   Parent 2: ggll

The gametes formed by each parent fly.

Parent 1: GL   Parent 2: gl

The punnett square for this cross:

		Parent 1 gametes
		GL
Parent 2 gametes	gl	GgLl

All offspring would have grey bodies and long wings

1b

b) The expected number of offspring showing the following phenotypes would be….

Calculations:

- Grey body, long wings: (4 800 ÷ 16) x 9
  AND
  Grey body, vestigial wings: (4 800 ÷ 16) x 3
  AND
  Ebony body, long wings: (4 800 ÷ 16) x 3
  AND
  Ebony body, vestigial wings: (4 800 ÷ 16) x 1; [1 mark]

Answers:

- Grey body, long wings: 2 700
  AND

Grey body, vestigial wings: 900

AND

Ebony body, long wings: 900

AND

Ebony body, vestigial wings: 300; [1 mark]

Full marks awarded for the correct answers only.

[Total: 2 marks]

According to Mendel, the expected Phenotypic ratios for a dihybrid cross between individuals heterozygous for both characteristics is:

$$9:3:3:1$$

Therefore:

- $\frac{9}{16}$ would be dominant for both characteristics → $(4\,800 \div 16) \times 9 = \underline{2700}$

- $\frac{3}{16}$ would be dominant for one and recessive for the other characteristic → $(4\,800 \div 16) \times 3 = \underline{900}$
  → $(4800 \div 16) \times 3 = \underline{900}$

- $\frac{1}{16}$ would be recessive for both characteristics → $(4800 \div 16) \times 1 = \underline{300}$ [1 mark]   [1 mark]

1c

c) The completed χ^2 table:

Two marks awarded for the correct answer and **one** mark allocated if all   values were correctly calculated.

Phenotype of offspring	Observed (O)	Expected (E)	($O - E$)
Grey body, long wings	2 580	2 700	(2 580 - 2 700) = **-12**
Grey body, vestigial wings	900	900	(900 - 900) = **0**
Ebony body, long wings	1 010	900	(1 010 - 900) = **110**
Ebony body, vestigial wings	310	300	(310 - 300) = **10**

- $\chi^2 = 5.33 + 0 + 13.4 + 0.333$; [1 mark]
- $= 19.1$;; [2 marks]

Error carried forward would apply for calculations of expected number of offspring from part b).

Full marks awarded for the correct answer only.

[Total: 3 marks]

For these calculations, you would need to copy over the expected number of offspring that would display the different phenotypes from the previous question. From there you can calculate the necessary values to determine the value of χ^2.

1d

d) The conclusions that can be drawn from these results are:

Accept the reverse argument based on the students' calculations from part c)

- The probability (that the difference is due to chance) is less than 0.05 (for 3 degrees of freedom) **OR** the value for chi-squared is greater than 7.82; [1 mark]
- There is a significant difference between the observed and expected results; [1 mark]
- This difference is therefore not due to chance; [1 mark]

[Total: 3 marks]

When writing your conclusion do not forget to state the probability (generally always 0.05) and the degrees of freedom (v), this is one less than the number of classes / phenotypes (which was 4 - 1 = 3 in this case). You will be awarded the marks if you correctly explain the meaning of the χ^2 value you calculated, even if the value itself was incorrect.

- The difference is due to the chance if χ^2 is less than the critical value (at p = 0.05)
- The difference is not due to chance if χ^2 is more than the critical value (at p = 0.05)

2a

a) The name of the statistical test used is...

- Chi-squared / χ^2 (test); [1 mark]

The expected phenotypic ratio for the F2 generation is...

- 9 : 3 : 3 : 1; [1 mark]

[Total: 2 marks]

The chi-squared test is used because we are testing for a significant difference between **observed** and **expected** values.

The expected ratio is 9:3:3:1; this is a ratio that you should recognise, and is the expected result of a dihybrid cross with **two parents who are heterozygous** for **both traits**:

2b

b) The genetic diagram should contain the following information...

- Parent genotypes = GgRr **AND** ggrr; [1 mark]
- Gametes = GR Gr gR gr **AND** gr; [1 mark]
- Offspring genotypes = GgRr **AND** Ggrr **AND** ggRr **AND** ggrr; [1 mark]
- Offspring phenotypes = Grey body red eyes **AND** grey body brown eyes **AND** black body red eyes **AND** black body brown eyes; [1 mark]

[Total: 4 marks]

Avoid wasting time drawing enormous Punnett squares when you only need a small one; this avoids repeating yourself when drawing Punnett squares containing parents that can only produce one or two types of gametes.

2c

c) These results are different from the expected results because...

Any **five** of the following:

- The expected ratio was 1:1:1:1 / the ratio is not 1:1:1:1; [1 mark]
- The flies are mostly grey(body)-red(eye) and black(body)-brown(eye); [1 mark]
- The majority resemble (original) parents / have parental phenotypes; [1 mark]
- There is a 1:1 ratio of two parental types **AND** a 1:1 ratio of recombinant types; [1 mark]
- Genes (for eye colour and body colour) are linked / on same chromosome; [1 mark]
- (alleles) G+R linked and g+r linked (in F1 / heterozygotes); [1 mark]
- No random/independent assortment (because the alleles are linked they are always inherited together); [1 mark]
- Crossing over (produces recombinants / minority phenotypes); [1 mark]
- Low numbers of recombinants shows that genes/loci are close together; [1 mark]

[Total: 5 marks]

It is worth bearing in mind that when offspring ratios do not match the expected ratios it is often due to the effects of **linkage**.

When genes are linked they are found at **different loci on the same chromosome**. This means that when chromosomes are separated during meiosis I the alleles at these loci are much more likely to be **inherited together**. This leads to offspring that have the same traits as their parents.

Any offspring that have a combination of traits that should not theoretically be possible (given the linkage of the genes) must have come about as a result of **crossing over**; alleles are exchanged, producing new allele combinations. The closer together the linked genes are the less likely they are to be recombined during crossing over, so a small number of recombinant individuals indicates close proximity of genes on a chromosome.

3a

a) A genetic diagram of a cross between two individuals who are heterozygous at the *PINK1* gene locus should contain the following...

- Symbols = normal (allele) = E **AND** EOPD (allele) = e; [1 mark]
- Parent genotypes and gametes = Ee x Ee **AND** E e E e; [1 mark]
- Offspring genotypes = EE Ee (x2) ee; [1 mark]
- Ratio = 3 normal : 1 EOPD; [1 mark]

Gametes and offspring genotypes can be shown within a Punnett square.

Accept alternative symbol choices, e.g. Pp for the PINK1 gene, provided that upper and lower case letters are clearly distinguishable and the key is clearly labelled.

[Total: 4 marks]

It is likely that symbols other than Ee would be accepted here provided that the upper case letter represents the dominant allele and the matching lower case letter represents the recessive allele. It's a good idea to choose letters where the upper case letter is distinctly different to the lower case letter to prevent misinterpretation, eg. choosing P and p here might mean that it is difficult for the examiner to tell the difference between a large P and a small p.

Be careful not to confuse genotype and phenotype here; the genotype is the combination of alleles that an individual has while the phenotype is the observable characteristics. The genotype ratio would be 1 EE : 2 Ee : 1 ee, but the phenotype ratio is 3 normal : 1 EOPD.

a)   Draw a genetic diagram of a cross between two individuals who are heterozygous at the
     PINK1 gene locus.

Include the following:

- key to symbols used for alleles
- parental genotypes
- gametes
- offspring genotypes
- ratio of offspring phenotypes

You MUST include everything that the question tells you to include!

pick symbols and give a clear key:

   E = dominant allele = normal
   e = necessive allele = EOPD   [1 mark]

Work out parent genotypes:

   We know that the parents are heterozygous
   So :

          Ee × Ee   [1 mark]

Show the parent gametes:

          Ⓔ ⓔ Ⓔ ⓔ   *Circles can be used to clearly distinguish gametes from genotypes*

Complete a Punnet square:

	E	e
E	EE	Ee
e	Ee	ee

Gametes can also be credited in Punnett square

offspring genotypes in Punnett square   [1 mark]

Give phenotype ratios:

   3 normal : 1 EOPD   [1 mark]

3b

b) A base substitution mutation can lead to the production of a non-functioning protein
kinase as follows...

Any **five** of the following:

- A <u>missense</u> mutation could occur; [1 mark]
- (This is when) the mRNA produced contains a different codon/triplet; [1 mark]
- A different tRNA binds (to the altered mRNA codon); [1 mark]
- A different amino acid is inserted / the primary structure (of the protein) is altered; [1 mark]
- The tertiary/3D structure (of the protein) is altered; [1 mark]
- (The change) may affect the active site (of the enzyme); [1 mark]
- (**OR**) a <u>nonsense</u> mutation could occur; [1 mark]
- (This results in a) stop codon; [1 mark]
- There is no matching tRNA / amino acid (for the stop codon); [1 mark]
- The protein is shortened / incomplete; [1 mark]

[Total: 5 marks]

Make sure that everything included in your answer is relevant to the question. For example no credit will be given here for points relating to silent mutations (which would still result in a functioning protein kinase) or to points relating to frame shift mutations (which do not occur as a result of base substitutions).

3c

c) This mutation causes EOPD in a heterozygote as follows…

Any **two** of the following:

- Only one (copy of the) allele is needed to produce the inhibitor; [1 mark]
- Other proteins cannot be phosphorylated/activated (by protein kinase A); [1 mark]
- Less aerobic respiration / ATP produced in (mitochondria of) neurones; [1 mark]
- (Lack of ATP leads to) inability to recycle dopamine / non-functioning sodium-potatssium pumps / inability to reach resting potential/repolarise neurone; [1 mark]
- Neurones die; [1 mark]

[Total: 2 marks]

The points in the mark scheme here are taken from several parts of the specification:

- You should understand the significance of the mutation being **dominant**
- You need to know the role of **active protein kinase A** in cell signalling; you will have come across this when learning about the role of glucagon in controlling blood glucose
- Consider the **role of mitochondria** (see part b) in the normal functioning of neurones

Only **one copy** of the dominant mutated allele needs to be present in order for it to affect the phenotype by producing an inhibitor protein. The inhibition of protein kinase A means that it can **no longer phosphorylate** another protein, and the enzyme cascade that it is part of will no longer occur. We know from part b) that this affects mitochondrial function; if mitochondria are not functioning then **ATP will not be produced** by aerobic respiration, and ATP is needed for several **processes inside nerve cells**, e.g. sodium potassium pumps to establish and re-establish resting potential, and the recycling of neurotransmitters.