Q1.

4 (a) metaphase;
II; (*allow one mark for telophase and two marks for telophase 1*)

2

(b) ref. spindles/microtubules shorten contract/pull/breakdown;
centromeres divide;
choromatids (pulled) apart;
to opposite poles;
chromosomes unwind/AW;
nuclear membrane reforms;
ref. cytokinesis/cleavage;

4 max

(c) independent/random assortment;
of homologous chromosomes;
different combinations of parental chromosomes;
crossing over/chiasmata;
between chromatids of homologous chromosomes/non-sister chromatids;
breaks up linkage groups/mixes alleles from parents; R genes
ref. to non-identical/genetically different gametes;

4 max

Total: 10

Q2.

5 (a) phenotype is the feature/characteristic;
results from interaction of genotype and environment on organism/ environment may alter the appearance of an organism;
genotype unaffected by environment;
genetic characteristics inherited/passed on to offspring/ora/represents alleles possessed;

2 max

(b) artificial selection carried out by humans;
choose organisms with useful characteristics/benefit to humans;
natural selection carried out by environment;
ref. survival (to breed);
ref. evolution;

3 max

(c) (i) length of DNA/sequence of bases/locus on a chromosome;
coding for a characteristic/protein/polypeptide/enzyme;

2

(ii) alternative form of a gene;
determining contrasting characters/controls one form of a character;
occupies same locus;
ref. sequence of bases;
ref. dominance;

3 max

Total: 10

Q3.

Question 5

(a)

black female	X	orange male
X^BX^B		X^OY ;
tortoiseshell female		black male
X^BX^O		X^BY ; *
orange female	X	black male
X^OX^O		X^BY ;
tortoiseshell female		orange male
X^BX^O		X^OY ; *

(* *must also have either gametes / construction lines / punnet square*).

(b)

tortoiseshell female

X^BX^O;

black female

X^BX^B ;

black male

X^BY ;

orange male

X^OY ;

(*phenotypes and genotypes must be linked otherwise max 2*).
(*penalize* ***once*** *for lack of gender*).

(b)

tortoiseshell female

X^BX^O ;

black female

X^BX^B ;

black male

X^BY ;

orange male

X^OY ;

(*phenotypes and genotypes must be linked otherwise max 2*).
(*penalize* ***once*** *for lack of gender*).

(c) X chromosome inactivated randomly early in development / AVP ;

1

Total : 9

Q4.

Question				Marks
2	(a)		metaphase 1 / (late) prophase 1 ; **R** early / middle	1
	(b)	1	ref. (homologous chromosomes) pairing / synapsis ;	
		2	ref. to chiasma / crossing over ;	
		3	exchange of genetic material ;	
		4	between non-sister chromatids / AW ;	3 max
	(c)	1	breakage of linkage groups / ref. new linkage groups ;	
		2	may have different alleles ;	
		3	creates new combinations of alleles ;	
		4	when sister chromatids separate ;	2 max
	(d)		ref. idea of random orientation at metaphase I and II / random alignment of homologous chromosomes on spindle equator ;	
			subsequently leads to independent assortment ;	
			2^n possible combinations when n is number of chromosome pairs ;	
			ref. to chromosome mutation qualified ;	
			extra detail ;	
			ref. gametes haploid (so can fuse) ;	
			random fusion of gametes ;	
			N.B. 3 sets of 2/3 marks	4 max
				Total: 10

Q5.

5	(a)		parental genotypes ; e.g. AaBb x AaBb gametes ; correct use of punnett square ; F1 genotypes ; F1 phenotypes ; (must link to genotypes) yellow and sphere $^1/_{16}$;	[6]
	(b)	(i)	contract / die from, malaria ;	[1]
		(ii)	contract / die from, sickle-cell anaemia ;	[1]
	(c)		resistant to malaria ; detail ; more likely to survive ; and reproduce ; pass on sickle-cell allele ;	[3 max]
				[Total: 11]

Q6.

7 (a) both alleles, influence phenotype / are expressed ;
ref. more than 2 phenotypes possible ;
phenotype of heterozygote different from either homozygote ; [3]

(b) son receives Y chromosome from father ;
Y chromosome does not carry haemophilia allele ;
father will pass haemophilia allele to daughter(s) ;
daughter will be, a carrier / heterozygous / X^HX^h ;
daughter may pass allele to, her son / his grandson ; *accept on diagram* [3 max]

(c) (i) (male) $C^BC^BX^aX^a$; x (female) $C^WC^WX^AY$;

(gametes) C^BX^a C^WX^A or C^WY ;

$C^BC^WX^AX^a$; (male, blue, barred)

$C^BC^WX^aY$; (female, blue, non-barred)

accept other symbols but only with key
if male XY and female XX then mark gametes and offspring genotypes to max 2
if other symbols used but no key then mark to max 2 [5]

(ii) blue colour is heterozygous / C^BC^W ;
test cross ;
with non-barred female ;
if all offspring barred, must be X^AX^A / homozygous ;
if some offspring non-barred, must be X^AX^a / heterozygous ; [3 max]

[Total: 14]

Q7.

6 (a) *allele*
(different) form of a gene ; **A** variety / version
ignore refs to locus / mutation [1]

recessive
allele which does **not** have its effect in heterozygote / allele which (only) has its effect in homozygote / affects phenotype if dominant allele is absent ; [1]

(b) gene / allele, on X chromosome / sex linkage ;

female, needs 2 RGC alleles / homozygous recessive / can be heterozygous ;

male needs 1 RGC allele ; [2 max]

(c) **1** – X^RX^r / **Rr** ;

4 – X^RY / **R** / R^o / **R-** ;

6 – X^rY / **r** / r^o / **r-** ;

7 – X^RX^r / **Rr** ; [4]

if X and Y not used then mark to max 3

[Total:8]

Q8.

7 **(a)** key ; *black upper case, chestnut lower case*

gametes ;

offspring genotypes **and** chestnut identified ;

25% / 0.25 / ¼ / 1 in 4, (probability) ; *ignore ratios* [4]

(b)

parental genotype	$aaCC^{CR}$		AaCC	
parental phenotype	palomino / cream		black ;	
gametes	**aC**	**aC^{CR}**	**AC**	**aC ;**
offspring genotypes	**AaCC**	**aaCC**	**AaCCCR**	**$aaCC^{CR}$;**
			any order	
offspring phenotypes	**black**	**chestnut**	**black**	**palomino / cream ;**
			order linked to genotype order	

ecf can be applied to offspring genotypes and phenotypes [4]

[Total: 8]

Q9.

6 **(a)** change in, DNA/base sequence ;
produces different allele ;
ref. different, protein/polypeptide, produced ; [2 max]

(b) 1 – X^rX^r ;
3 – X^rY ;
9 – X^RX^r ;
10 – X^RY ; [4]

(c) *answers must refer to phosphate ions*

1 altered shape/non-functional/no, carrier protein ;

2 less/no, reabsorption of phosphate ions (into blood) ;

3 from, glomerular filtrate/lumen of/proximal convoluted tubule ;

4 more/all, phosphate ions excreted ;

5 low phosphate ion concentration in, blood/bones ; **R** no phosphate ion conc [2 max]

[Total: 8]

Q10.

6 **(a)** 1 allele/gene, found on **X** chromosome ;

2 females have two copies of, allele/gene ;

3 males have only one copy of, allele/gene ; [2 max]

(b) *key to symbols*

recessive allele X^a (= allele for CI)

dominant allele X^A (= allele for normal iris) ;

cross 1
parental phenotypes male with CI/cleft iris **and** normal female ;

gametes X^a **or** Y **all** X^A ;

offspring genotypes X^AX^a X^AY ;

offspring phenotypes **normal female** **normal male** ;

.....

or

.....

cross 2
parental phenotypes male with CI/cleft iris **and** normal female ;

gametes X^a **or** Y X^A **or** X^a ;

offspring genotypes X^AX^a X^AY X^aX^a X^aY ;

offspring phenotypes **normal female** **normal male** **cleft iris/CI female** **cleft iris/CI male** ; [5]

offspring phenotypes must be linked to genotypes

(c) 1 in 4/25%/0.25 ; **R** ratios [1]

[Total: 8]

Q11.

6 **(a)** enzyme acts on only one substrate ;
shape of active site is complementary to substrate ;
AVP ; e.g. substrate held by temporary bonds / ES complex [2 max]

(b) symbols (must be of same letter) ;
parental genotypes **and** gametes ;
offspring genotypes **and** phenotypes **linked** ; [3]

(c) 1. insulates axon (membrane) ;
2. depolarisation occurs only at nodes (of Ranvier) / AW ;
3. local circuits ;
4. saltatory conduction / AW ;
5. speeds transmission of, action potential / impulse ;
6. AVP ; e.g. speed increases up to 50 times / 100ms^{-1} [3 max]

[Total: 8]

Q12.

7 **(a)**

nuclear division	letter of stage
meiosis I	B
	E
	J
	H
	F
	D
meiosis II	G
	I
	C
	A

E J H F all in meiosis I ;
E J H F in correct order ;
G I C A all in meiosis II ;
G I C A in correct order ; [4]

(b) 1. chiasma / crossing over ;
2. between non-sister chromatids ;
3. homologous chromosomes / bivalents ; *in correct context of mp1 or mp8*
4. in prophase I ;
5. exchange of genetic material / AW ;
6. linkage groups broken ;
7. new combination of alleles ;
8. independent assortment ; **R** random assortment
9. in metaphase I ;
10. detail of independent assortment ;
11. AVP ; e.g. possible mutation [5 max]

[Total: 9]

Q13.

9 **(a)** *gene mutation*

1. spontaneous / random, change ;
2. in, base sequence / nucleotide sequence / mRNA code / codon ;
3. example ; e.g. addition / insertion / substitution / deletion / inversion

triplet code

4. (sequence of) three (DNA nucleotide) bases ;
5. complementary to mRNA codon ;
6. codes for a specific amino acid ; 4 max

(b)

parental phenotypes	man without HD	woman with HD
parental genotypes	**tt**	**Tt**
gametes	**all t**	**T or t ;**
offspring genotypes	**Tt**	**tt**
offspring phenotypes	**Huntington's disease**	**normal ;**

probability of first child having D **50% / 0.50 / 1 in 2 ;** [3]

[Total: 7]

Q14.

7 **(a)** correct symbols ; e.g. X^A = (allele for) red-eye
X^a = (allele for) white-eye

parental genotypes $X^A X^a$ and $X^a Y$;

gametes X^A X^a X^a Y ;

offspring genotypes $X^A X^a$ $X^A Y$ $X^a X^a$ $X^a Y$;

offspring phenotypes red-eyed female, red-eyed male, white-eyed female, white-eyed male ; [5]

(b) (i) passes **Y** chromosome onto son / passes **X** chromosome onto daughter ; [1]

(ii) heterozygous ; [1]

(iii) gene / allele, mutation ; [1]

[Total: 8]

Q15.

7 (a) *heterozygous*
two different alleles of a gene / different allele pair for a gene / AW ;

produces gametes with different genotypes ; *max 1*

genotype
alleles present in an organism / particular alleles of a gene / genetic constitution / AW ; [2]

(b) *parental genotypes*
AaDd x AaDd ;

gametes
AD Ad aD ad x AD Ad aD ad ;

two marks for correct Punnett square ;; *deduct one mark for each mistake*

(all 4) phenotypes linked correctly to genotypes ;

(probability of yellow offspring) 3 out of 16 **or** 0.19 **or** 19% ; [6]

[Total: 8]

Q16.

7 (a) *sex-linked*
(gene) carried on, one sex chromosome/**X,** and not on, the other/**Y;**

gene
section of DNA/sequence of nucleotides/sequence of bases,
that codes for a (particular) polypeptide; [2]

(b)

parental phenotypes	*tortoiseshell female*		*black male*	
parental genotypes	X^BX^O		X^BY;	
gametes	X^B	X^O	X^B	Y;
offspring genotypes	X^BX^B	X^BY	X^BX^O	X^OY;
offspring phenotypes	**black female**	**black male**	**tortoiseshell female**	**orange male;**

[4]

(c) tortoiseshell is heterozygous;

males, heterogametic/only one **X** chromosome;

(therefore) only one copy of gene/only black or orange allele present; [max 2]

[Total: 8]

Q17.

7 (a) W^R = allele for warfarin resistance
W^S = allele for warfarin susceptibility

parental phenotypes	resistant male		resistant female	
parental genotypes	$W^R W^S$		$W^R W^S$	
gametes	W^R	W^S	W^R	W^S ;
offspring genotypes	$W^R W^R$	$W^R W^S$	$W^R W^S$	$W^S W^S$;
offspring phenotypes	**resistant**	**resistant**	**resistant**	**susceptible** ;

[3]

(b) not enough Vitamin K found (in the wild) / require too much Vitamin K; [1]

(c) competitive / reversible;

as the concentration of inhibitor increases, the rate of the (inhibited) reaction decreases
or
as dose of warfarin increases, the rate at which blood clots decreases; **ora** [2]

(d) 1. different, codon / triplet;
2. stop codon;
3. different amino acid;
4. different, primary / secondary / tertiary / 3D, structure;
5. shortened, polypeptide / protein;
6. change in function of protein; [3 max]

[Total: 9]

Q18.

Question 4

(a)

metaphase ; **1**

(b)

centromeres divide / splits; **R** break
chromatids separate ;
idea movt. to opposite poles / centrioles ;
by microtubules / spindle fibres ;
idea.mechanism of movement ; **3 max**

(c)

(i)

breaks down / disperses ; **1**

(ii)

centrioles divides/replicate;
to form two pairs (of centrioles) ;
move to (opposite) poles; **2 max**

(d)

1 random alignment / independent assortment / or description;
different mix of maternal and paternal chromosomes/chromatids ;

2 crossing over / chiasmata formation/exchange of genetic material ;
between chromatids of homologous chromosomes ;
breaks up linkage groups / mixes maternal and paternal alleles ;

In 1 or 2 ref. different gametes produced; **4 max**

Total : 11

Q19.

Question 5
(a)
Either
If genetic diagram used **Penalise once for incorrect symbols**

orange dominant to black (or converse);

orange scallop

parents	$S^o S^b$	X	$S^o S^b$		;
gametes	S^o S^b		S^o S^b		;
genotype	$S^o S^o$	$S^o S^b$	$S^o S^b$	$S^b S^b$	
phenotype	orange			black	;

black scallop

parents	$S^b S^b$	X	$S^b S^b$	;
gametes	(	S^b	S^b)	
genotype		$S^b S^b$		
phenotype		black		;

Or
If text explanation given

orange dominant to black (or converse);
orange are heterozygous;
(because) ref. 3:1 ratio;
link data to ratio;
black are homozygous;
because all offspring are black; **6**

(b)
separate orange scallops produced from first cross / test cross orange with black ;
some will produce only orange offspring ;
these will be homozygous for orange allele/pure breeding ; **2 max**

Total : 8

Q20.

Question 4

(a) parental genotype ;
gametes ;
offspring genotype ;
offspring phenotype ;
penalise once if other symbols used — 4

(b) suffer from vitamin K deficiency / require too much vitamin K ; — 1

(c) warfarin will kill rats without resistance - homozygous recessive ;
homozygous dominant rats require too much vitamin K ;
heterozygous rats most likely to survive and produce offspring ;
only 50% of offspring will be heterozygous ; — 3 max

(d) results in a different codon / triplet ;
(may) result in change of amino acid ;
different primary protein structure ;
this may result in change in protein function ;
suitable example e.g. sickle cell anaemia ; — 3 max

Total : 11

Q21.

Q22.

Question 2

(a) correct parental genotypes ;
correct gametes ;
correct genotypes of offspring ;
correct phenotypes linked to genotypes ; [4]

(b) yellow shrunken homozygous ;
double recessive ; [2]

(c) (381 x 3/16) = 71 (36/71) = 0.507;
(381 x 1/16) = 24 (9/24) = 0.375 ;
1.80 ; [3]

(d) greater than 0.5 ; allow ecf [1]

(e) difference from expected not significant ; allow ecf
because greater than 0.5 ;
ratio phenotype is 9:3:3:1 ;
(the small) observed differences are due to chance ; [2 max]

Total [12]

Q23.

Question			Expected Answers		Marks
2	(a)	(i)	black : red ; 1 : 1 ;		2
		(ii)	black : copper : red ; 2 : 1 : 1 ;		2
		(iii)	red copper ; 3 1 ;		2
	(b)	(i)	test / back, cross ; with, copper / A^tA^t / homozygous recessive ;		
		(ii)	if all offspring red, homozygous ; if some offspring copper, heterozygous ; ref. equal proportions of offspring ;	*mark (i) and (ii) together*	4 max
				[Total: 10]	

Q24.

3 (a) *any four from*

1 thick / dehydrated / sticky, mucus ;
2 builds up in, lung / gut / airways ; ***A*** *excess of mucus…..* ***R*** *blocks up*
3 infections in lungs ; ***A*** *named infection*
4 scar / damage, lungs ;
5 mucus, prevents secretion (of digestive enzymes) from pancreas / blocks pancreatic duct ;
6 malnutrition / inadequate digestion / inadequate absorption ; ***R*** *indigestion*
7 reduced, growth / development ;
8 excessively salty sweat / muscle cramps ;
9 mucus blocks sperm duct / males sterile ; *female neutral* [4 max]

(b) *gametes* BX bX BX BY bX bY ;

offspring genotypes see table ;

offspring phenotypes see table ; ***R*** *phenotypes if no gender*

probability of CF daughter 1in 8 offspring / 1 in 4 daughters / 12.5% / 0.125 ;

gametes	BX	BY	bX	bY
BX	BBXX normal female	BBXY normal male	BbXX normal/carrier female	BbXY normal/carrier male
bX	BbXX normal/carrier female	BbXY normal/carrier male	bbXX CF female	bbXY CF male

[4]

(c) 1 mutation alters DNA base sequence ;
2 triplet of bases / three bases,(in DNA) codes for an amino acid ; ***R*** *'codon' re DNA*
3 base substitution alters code ;
4 base, addition / deletion, produces frame shift / subsequent triplets have altered coding ;
5 ref. transcription ;
6 ref. translation ; [4 max]

(d) **(i)** E has, AAG / GAA / 2As and 1G, missing / ora ; [1]

(ii) E's polypeptide lacks one amino acid present in D's ;
different primary structure ;
may have different, secondary structure / tertiary structure / 3D shape ; [2 max]

[Total: 15]

Q25.

8 1 CC^a Bb X C^hC^a Bb ;

2 CB Cb C^aB C^ab x C^hB C^hb C^aB C^ab ;

3 offspring phenotypes:
full black : full red : himalayan black : himalayan red : albino black : albino red ;

4 phenotype ratio:
6 : 2 : 3 : 1 : 3 : 1 ;

5/6 offspring genotypes in Punnett square ;; [6]

ecf
if incorrect symbols penalise the parent genotypes (pt 1) and mark rest of cross up to max 4

ecf
if one gene only used then mark to max 2

[Total: 6]

Q26.

				[Total: 7]
7	**(a)**		*allele* different / alternative, form of a gene ; **A** variety of a gene *dominant* (allele) that always expresses itself in the phenotype when present / (allele) which influences the phenotype even in the presence of an alternative allele / AW ;	[2]
	(b)		parental phenotype ; e.g. striped / long x striped / long **A** wild x wild parental genotype ; e.g. AaBb x AaBb gametes ; e.g. AB Ab aB ab offspring genotypes ;; offspring phenotypes ; *must be linked to genotypes*	[6]
			accept other symbols if key used *penalise once for no key but only if genetic cross works*	

	(c)	(i)	<table><tr><th></th><th colspan="4">phenotypes of *Drosophila melanogaster*</th></tr><tr><th></th><th>grey body long wing</th><th>grey body vestigial wing</th><th>ebony body long wing</th><th>ebony body vestigial wing</th></tr><tr><td>observed number (O)</td><td>207</td><td>79</td><td>68</td><td>30</td></tr><tr><td>expected ratio</td><td>9</td><td>3</td><td>3</td><td>1</td></tr><tr><td>expected number (E)</td><td>216</td><td>72</td><td>72</td><td>24</td></tr><tr><td>$O - E$</td><td>–9</td><td>7</td><td>–4</td><td>6</td></tr><tr><td>$(O - E)^2$</td><td>81</td><td>49</td><td>16</td><td>36</td></tr><tr><td>$\frac{(O-E)^2}{E}$</td><td>0.38</td><td>0.68</td><td>0.22</td><td>1.50</td></tr></table>	[3]
		(ii)	2.78 ; *apply ecf*	[1]
		(iii)	χ^2 value represents probability of > 0.05 ; no significant difference ; (probability shows) differences due to chance ;	[2 max]
				[Total:14]

Q27.

7	(a)		parents, carriers / heterozygous ; child homozygous recessive ; ¼ / 0.25 / 25%, chance ; mutation ;	[3 max]
	(b)	(i)	gene technology / genetic engineering / description ;	[1]
		(ii)	glucagon ;	[1]
		(iii)	low blood glucose concentration / during or after exercise ; **R** sugar	[1]
	(c)		foreign / non-self / cell recognition ; stimulates immune response / AW ;	[1 max]
	(d)		*parental genotypes* L^ML^N x L^ML^N *gametes* L^M or L^N L^M or L^N ; *parental genotypes and gametes for one mark* *offspring genotypes* L^ML^M L^ML^N L^ML^N L^NL^N ; *offspring phenotypes* MM MN MN NN ;	[3]
			penalise once for omission of L	

	(e)		Canadian Inuit, allele frequencies / L^ML^N ratio, different from others ; high frequency of L^M / low frequency of L^N , compared to other populations ; **R** just highest L^M / lowest L^N less outbreeding / more inbreeding ; AVP; e.g. L^M has selective advantage in Inuit environment	[3 max]
				[Total: 13]

Q28.

6 (a) variation / different form, of a gene ; [1]

(b) *marks for reasons only*
$Hb^A Hb^A$
low – susceptible to / die from, malaria ;

$Hb^A Hb^S$
high – no (full blown) SCA / have SC trait ;
not, susceptible to / likely to die from, malaria ;

$Hb^S Hb^S$
low – susceptible to / die from, SCA ; [4]

(c) 1 *USA* malaria not selection pressure ;

2 Hb^S no advantage ;

3 due to outbreeding ;

4 genetic testing can lead to termination of pregnancy **or** testing / counselling, leads to not having children ; [2 max]

[Total: 7]

Q29.

9 **(a)** *dominant*
(allele) that always expresses itself (in the phenotype) when present
or
(allele) which influences the phenotype even in the presence of an alternative allele ;

gene
length of DNA / sequence of nucleotides, coding for a (specific) polypeptide ; **A** protein [2]

(b)

parental phenotypes	man without TSC	woman with TSC	
parental genotypes	**tt**	**Tt**	
gametes	**all t**	**T or t**	;
offspring genotypes	**Tt**	**tt**	
offspring phenotypes	**TSC**	**normal**	;
probability of child having TSC	**50% / 0.50 / 1in 2 ;**		

[3]

(c) 1. spontaneous / random / chance ;
2. mutation of, gene / allele ;
3. AVP ; e.g. named mutagen / detail of mutation [2 max]

[Total: 7]

Q30.

6 **(a)** *allele*
different / alternative, form of a gene ; **A** variety of a gene

one of two or more alternative nucleotide sequences at a single gene locus ; [1 max]

dominant
(allele) that (always) expresses itself in the phenotype when present /
(allele) which influences the phenotype even in the presence of an alternative allele ; [2]

(b) parental genotypes ;
gametes ;
offspring genotypes (in Punnett square) ;;
offspring phenotypes linked to genotypes ;
ratio 9:3:3:1 linked to phenotypes ; [6]

[Total: 8]

Q31.

6 **(a)** **(i)** *accept answers in a genetic diagram where genotypes are linked to phenotypes*
1 agouti allele / C^a, dominant to black allele / C^b ; **ora**
2 black parents homozygous recessive ;
3 agouti parents heterozygous **or** homozygous ; [2 max]

(ii) *accept answers in a genetic diagram where genotypes are linked to phenotypes*
1 yellow allele / C^y, dominant to, black allele / C^b ;
2 ref. to modified 3:1 ;
3 (homozygous) genotype $C^y C^y$, lethal / does not survive ; [2 max]

(iii) *accept answers in a genetic diagram where genotypes are linked to phenotypes*
1 yellow allele / C^y, dominant to **all** others ;
2 agouti / C^a **or** black and tan / C^{bt}, allele, dominant to black allele ;
A black allele recessive to all other alleles
3 yellow mice all heterozygous (must be stated) ; [2 max]

(b) 1 cross (black and tan mouse) with, black mouse / homozygous recessive mouse / $C^b C^b$;
2 if **all** offspring black and tan then parent, $C^{bt} C^{bt}$ / homozygous ;
3 if some offspring are black (and some are black and tan) then parent, $C^{bt}C^*$ / heterozygous ; [2 max]

[Total: 8]

Q32.

1 **(a)** allele – variation / different form, of a gene ;

dominant – (allele) always expresses itself (in the phenotype when present) ; [2]

(b) the greater the number of (CAG) repeats the earlier the symptoms first appear / inversely proportional / negative correlation ;

paired figures ; [2]

(c) 1. fear of needles ;

2. fear of positive result ;

3. fear of effect of result on other members of family ;

4. no desire to have children ;

5. financial / insurance, concerns / AW ;

6. possibility of false results ;

7. cost of test ;

8. not worth having test because of no treatment ; [max 3]

[Total: 7]

Q33.

1 (a) X^RY and X^rX^r ;

X^R Y X^r (X^r) ; *allow ecf from incorrect parental genotypes*

X^RX^r and X^rY ; [3]

(b) (i)

phenotype of fly	O	E	O–E	$(O–E)^2$	$\frac{(O–E)^2}{E}$
red-eyed female	54	50	(+)4	16	0.32 ;
white-eyed male	46	50	(-)4	16	0.32 ;

0.64 ;
allow ecf [3]

(ii) probability is greater than 0.05 ; **A** chi squared smaller than 3.84

no significant difference ;

due to chance ; [max 2]

[Total: 8]

Q34.

7 (a) centromere ; [1]

(b) *idea that* different genes, are present / missing ; **R** alleles

different, proteins / poypeptides, produced / missing ; [2]

(c) XY ;

X X_1 X Y ;

XX XX_1 ;

normal Turner's ; [4]

[Total:7]

Q35.

7 **(a)** symbols and key ; e.g. A = NF allele and a = normal allele

parental genotypes **and** gametes ; e.g. parental genotypes Aa x aa
gametes A a x a a

offspring genotypes **and** phenotypes **linked** ; e.g. Aa has NF and aa is unaffected [3]

(b) spontaneous / random / chance ;

mutation of, gene / allele ;

AVP ; e.g. named mutagen / detail of mutation / in oocyte / in sperm [max 2]

(c) compresses nerve ;

damages, myelin sheaths / Schwann cells ;

prevents, setting up of local circuits / saltatory conduction ;

stops Na^+ / K^+ pumps from working ;

blocks blood supply;

qualified ; e.g. effect on, oxygen supply / glucose supply / ATP production

AVP ; e.g. may stop ion channels opening [max 3]

[Total:8]

Q36.

7 **(a)** *recessive*
only expressed in homozygote/two copies of the allele needed to be expressed/
not expressed in heterozygote/not expressed in presence of dominant allele ;

mutation
change in the structure of, DNA/gene/allele
or
change in, base/nucleotide, sequence ; [2]

(b) suitable symbols and key ; e.g. A = allele for normal (non PKU)
a = allele for PKU

correct parental genotypes **plus** correct gametes ;

offspring phenotypes linked to correct offspring genotypes ; [3]

(c) 1 fewer amino acids ;

2 change in primary structure ; **A** different amino acid sequence

3 different, tertiary structure/3D shape ;

4 *ref. to* active site of, PAH/enzyme, changed/absent ;

5 PAH/enzyme/protein, non-functional/AW ; **A** different function [max 3]

[Total: 8]

Q37.

7 **(a)** *gene*
length/section, of DNA
or
sequence of, bases/nucleotides ;

coding for a, polypeptide/protein ;

allele
different/alternative, form of a gene ; **A** variety of a gene

occupying same, locus/position (on homologous chromosomes) ; [4]

(b)

individual	phenotype	genotype	
1	B	$I^B I^o$	;
2	A or B	$I^A I^o$ or $I^B I^o$	;
3	B or A	$I^B I^o$ or $I^A I^o$	;
4	A	$I^A I^o$	;

Individuals 2 and 3 ***must*** *have different phenotypes and genotypes* [4]

[Total: 8]

Section_B

1.

9 (a) *do not credit marking points out of sequence*

prophase 1

1 idea of condensation of chromosomes ;

2 homologous chromosomes pair up / bivalent formed ;

metaphase 1

3 homologous chromosomes / bivalents, line up on equator ;

4 of spindle ;

5 by centromeres ;

6 independent assortment / described ;

7 chiasmata / described ;

8 crossing over / described ;

anaphase 1

9 chromosomes move to poles ;

10 homologous chromosomes / bivalents, separate ;

11 pulled by microtubules ;

12 reduction division ;

metaphase 2

13 chromosomes line up on equator ;

14 of spindle ;

anaphase 2

15 centromeres divide ;

16 <u>chromatids</u> move to poles ;

17 pulled by microtubules ;

18 ref. haploid number ;

allow 4 ***or*** *14*
allow 11 ***or*** *17*

[9 max]

(b) 19 change in, base / nucleotide, sequence (in DNA) ;

20 during DNA replication ;

21 detail of change ; e.g. base, substitution / addition / deletion

22 frame shifts / AW ;

23 different / new, allele ;

24 random / spontaneous ;

25 mutagens ;

26 ionising radiation ;

27 UV radiation / mustard gas ; [6 max]

[Total: 15]

2.

11 (a) 1. (amino acid) code is three, bases / nucleotides ; **A** triplet code
2. (gene) mutation ; **R** chromosome mutation
3. base / nucleotide, substitution / addition / deletion
4. addition / deletion, has large effect (on amino acid sequence) ;
5. frame shift ;
6. completely new code after mutation / alters every 3 base sequence which follows ;
7. substitution may have little or no effect / silent mutation ;
8. different triplet but same amino acid / new amino acid in non-functional part of protein ;
9. substitution may have big effect (on amino acid sequence) ;
10. could produce 'stop' codon ;
11. sickle cell anaemia / PKU / cystic fibrosis ;
12. reference to transcription or translation in correct context ; **A** description [8 max]

(b) 13. (haemophilia) allele on X chromosome ; **A** gene
14. sex-linked ;
15. (haemophilia) allele recessive ;
16. man, homogametic / has one X chromosome ;
17. Y chromosome does not have blood clotting gene ;
18. only daughter(s) get his X chromosome ;
19. daughter(s) carrier(s) of (haemophilia) allele ;
20. grandson(s) 50% chance of having, (haemophilia) allele / haemophilia ;
21. granddaughter(s) 50% chance of carrying, (haemophilia) allele ;

allow following marks from diagram

22. correct symbols ; e.g. X^H and X^h explained
23. man's genotype ; e.g. X^hY *ignore partner's genotype*
24. F1 (daughter's) genotype ; e.g. X^HX^h *ignore her partner's genotype*
25. F2 (grandson's) genotypes ; e.g. X^hY X^HY *both required*
26. F2 (granddaughter's) genotypes ; e.g. X^HX^H X^HX^h *both required* **or** X^hX^h X^HX^h [7 max]

[Total: 15]

3.

9 **(a)** **1.** reduction division / (to) halve number of chromosomes / diploid to haploid / AW ;
2. homologous chromosomes pair up / bivalents form ;
3. ref. chiasmata / ref. crossing over ;
4. homologous chromosome pairs / bivalents, line up on equator ;
5. independent assortment ;
6. spindle / microtubules, attached to centromeres ;
7. chromosomes of each pair pulled to opposite poles ;
8. by shortening of, spindle / microtubules ;
9. nuclear envelopes re-form ;
10. cytokinesis / AW ; [6 max]

(b) *accept alternative symbols for alleles throughout*
11. frequency of sickle cell anaemia is highest in areas where malaria is common ;
12. sickle cell anaemia red blood cells cannot carry oxygen very well / AW ;
A sickling blocks capillaries
13. homozygous H^S / H^SH^S, have sickle cell anaemia / may die ;
14. homozygous H^N / H^NH^N, have normal, Hb / red blood cells ;
15. heterozygotes, have sickle cell trait
or
(sickle cell trait) red blood cells not (severely) affected ;
16. malaria parasite / *Plasmodium*, affects red blood cells ;
17. malaria lethal ;
18. sickle cell trait people / heterozygotes, less likely to suffer from (severe effects of) malaria ;
19. have selective advantage ;
20. pass on both H^N and H^S ;
21. malaria selects against, homozygous H^N / H^NH^N ;
22. sickle cell anaemia selects against, homozygous H^S / H^S H^S ;
23. *idea that* sickle cell allele is maintained within population
because of sickle cell trait individuals ; [9 max]

[Total: 15]