## Mark schemes

## 1

Parental genotypes: $\quad \mathrm{Gg} n \mathrm{nn} \quad \mathrm{gg} \mathrm{Nn}$;
Gamete genotypes

| Gn gn | gN |  |
| :---: | :---: | :---: |
| Gn | gN | gn |
| gn | Gg Nn <br> Grey, normal | Gg nn <br> Grey, vestigial |
|  | gg Nn <br> Ebony, normal | gg nn <br> Ebony, vestigial |

All offspring genotypes correct;
All offspring genotypes correctly derived;
[4] (a) (i) Only seen in males / not in females;
2
(ii) Unaffected parents / mother $\rightarrow$ child with M.D. /

$$
(1 \times) 2 \rightarrow 5 /(3 \times) 4 \rightarrow 11 / 8(\times 9) \rightarrow 13 ;
$$

$$
1 \text { (b) } 5=X^{d} Y
$$

$6=X^{D} Y$
$7=X^{D} X^{d} \underline{A N D} X^{D} X^{D}$
$8=X^{D} X^{d} ;$

$$
\begin{aligned}
& \text { All } 4 \text { correct = } 2 \text { marks } \\
& 2 \text { or } 3 \text { correct = } 1 \text { mark }
\end{aligned}
$$

$\max 2$
(c) $1 / 4 / 0.25 / 25 \% / 1: 3 / 1$ in $4 ;(N O T ‘ 1: 4 ’)$
] (a) Cannot make (active) enzyme A (which converts precursor to linamarin) / cannot make
(b) (i) $\mathbf{A L}+\mathbf{A I}+\mathbf{a L}+\mathbf{a l}$;
(d) From parental genotypes: AaLI $\times$ AaLI (no mark)
[Note: If wrong parental genotypes / wrong gametes: ALLOW correct derivation of offspring genotypes] (= max 1)

Correct derivation of offspring genotypes:

|  | AL | AI | aL | al |
| :--- | :--- | :--- | :--- | :--- |
| AL | AALL | AALI | AaLL | AaLI |
| AI | AALI | AAll | AaLI | Aall |
| aL | AaLL | AaLI | aaLL | aaLI |
| al | AaLI | Aall | aaLI | aall |

Correct identification of offspring genotypes with at least one A and two I alleles (= grey cells in above table);
Correct proportion: 3 / 16 / 3:13 / 18.75\% ;
(ii) The difference (from expected / from chance variation) is significant / difference / results not just due to chance;
Reject null hypothesis;
Being cyanogenic does help protect from slug damage;
(f) High slug population:

1. Find only cyanogenic plants / only cyanogenic plants survive;
2. (Cyanide release) limits / stops feeding by slugs / slugs killed;
[Accept: converse argument re. acyanogenic plants]

Low slug population:
3. Find both types of plant;
4. Less selection pressure from slugs / no selective advantage / no selection /described;
[15] (a) Gg / suitable equivalent;

Grey : black about 3: 1;
[Note: Can be in table / diagram]
2
(b) To determine the probability;
[Accept: Likelihood]
Of the results being due to chance;
[Accept: Coincidence]
(c) (i) both alleles will be expressed (in the phenotype);
(ii) $0.25 / 25 \%$; $=2$ marks
$C^{N}=250 / 1000 ;=1$ mark
(iii) $\quad P^{2}=(0.25)^{2} / 0.0625 /$ square of calculated figure for $\mathrm{C}^{\mathrm{N}}$; $=2$ marks $p^{2}+2 p q+q^{2}=$ 1.0; = 1 mark
$=31.25 / 31$;
[Accept: Derived from either $p^{2}$ or $q^{2}$ ]
[10]
(a) Only expressed in the homozygote / not expressed in the heterozygote / not expressed if

5 dominant present;


If parental genotype incorrect allow 1 mark for correct gametes based on given genotype and 1 mark for correct cross based on these gametes = 2 max MUST be clear link between F1 genotype and phenotype.
(c) (White) not camouflaged / not got stripes / white colour stands out;

Prey can take avoidance or are aware earlier / sooner;
Must have a time reference
2 max
[7] (a) (i) $B B X^{A} Y, B b X^{A} Y$;
6
(ii) $\quad \mathrm{Bb} X^{A} \mathrm{X}^{\mathrm{a}}, \quad \mathrm{bb} X^{A} \mathrm{X}^{\mathrm{a}}$;

1
(b) parental genotypes - $\mathrm{BbX}^{A} Y \quad \mathrm{x} \quad \mathrm{BbX}^{A} \mathrm{X}^{\mathrm{a}}$;

1

1

1
Genotypes of sons-

|  |  | Male gametes |  |
| :---: | :---: | :---: | :---: |
|  |  | BY | bY |
| Female gametes | $B X^{\text {A }}$ | $B^{\prime \prime}{ }^{\text {A }}$ Y | $\mathrm{BbX}^{\text {A }}$ Y |
|  | $B X^{\text {a }}$ | $B B X^{a} Y$ | $B b X^{\text {a }} \mathrm{Y}$ |
|  | $b X^{\text {A }}$ | $B b X^{A} Y$ | $\mathrm{bbX}^{\text {A }} \mathrm{Y}$ |
|  | $\mathrm{b}^{\text {a }}$ | $B b X^{\text {a }} \mathrm{Y}$ | bb $X^{\text {a }} \mathrm{Y}$ |

0.125 / 12.5\% / 1/8;

1
[6] (a) aabb;
(b) AaBb and aabb;
(c) Pea comb offspring will produce blue eggs;

Alleles $\mathbf{A}$ and $\mathbf{B}$ are inherited together / are on the same chromosome;
(d) Reference to crossing over;

Reduce chance of genes being separated (by crossing over);
If crossing over occurred some gametes will contain alleles $\mathbf{A}$ and $\mathbf{b}$;
2 max
(e) Two suitable environmental factors;
e.g.

Diet / named component of diet;
Temperature;
Light intensity / duration;
Disease;
2 max
(f) Cross $\mathrm{C} / \mathrm{X}^{f} \mathrm{X}^{\dagger}$ and $\mathrm{X}^{\mathrm{F}} \mathrm{Y}$;

1
(Only) cross where all males are one phenotype and all females are a different phenotype;
Cross showing all males are slow feather production, all females fast feather production;
(g) Two alleles for each gene present in male / chromosomes are homologous in male;

Female has one allele for each gene;
Recessive alleles always expressed in female;
Males need two recessive alleles for allele to be expressed / in males recessive alleles can be masked by dominant allele

3 max
[14] (a) hhDD, hhDd;
(both correct 1 mark)
(b) Epistasis;

One gene controlling / inhibiting the expression of another;
(c) Gametes correct HD, Hd, hD, hd, hd
(correct for both parents);
Genotypes HhDd, Hhdd, hhDd, hhdd ;
Phenotypes wiry wiry non-wiry, short non-wiry, long
$\begin{array}{llll}\text { Ratio } 2 & 1 & \\ \text {; }\end{array}$
(a) Two linked points:

Crossing over / exchange of material (between chromatids);
Different combinations of alleles / linkage groups changed / broken;
$O R$
Independent assortment / alignment of (homologous) chromosomes;
Different combinations of (maternal and paternal) chromosomes / alleles;
2 max
(b)

|  | D |  | $\|$d <br> Gamete genotype <br> Offspring genotype <br> Offspring Abnormal males / (all) | (no females); phenotypes |
| :--- | ---: | :--- | :--- | :--- |

3
[5] (a) (i) black;

## 10

(ii) chocolate;
(b) BE, Be, bE, be and be;

BbEe, Bbee, bbee, bbEe;
1 black: 2 yellow: 1 chocolate;
(c) (i) no enzyme coded for when no dominant / E allele; phaeomelanin not converted - (remains yellow);
(ii) E allele results in enzyme producing eumelanin;

B allele - more eumelanin deposited in hairs;
[9] (a) males are $X Y$ and females $X X$ / males have one $X$ chromosome and females two $X$
11 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;
(b) (i) $5-\mathrm{hh} \mathrm{X}^{b} \mathrm{Y}$; $6-\operatorname{Hh} X^{B} X^{b}$;
(ii) $\mathrm{h} \mathrm{X}^{\mathrm{b}}, \mathrm{h} \mathrm{Y}$, and $\mathrm{H} \mathrm{X}^{\mathrm{B}}, \mathrm{h} \mathrm{X}^{\mathrm{B}}, \mathrm{H} \mathrm{X} \mathrm{X}^{\mathrm{b}}, \mathrm{h} \mathrm{X}^{\mathrm{b}}$;
(iii) $1 / 8$ or $12.5 \%$ or 0.125 ;;
either genetic diagram to show genotypes $\mathrm{Hh} \mathrm{X}^{b} \mathrm{X}^{b}, \mathrm{Hh}^{\mathrm{B}} \mathrm{Y}$, hh $X^{B} X^{b}$, hh $X^{B} Y, H H X^{b} X^{b}, \operatorname{Hh}^{b} Y$, hh $X^{b} X^{b}$; hh $X^{b} Y$;
1/8;
or
$\mathrm{P}($ boy $)=0.5, \mathrm{P}($ colour blind $)=0.5, \mathrm{P}($ white streak $)=0.5$; ( $0.5 \times 0.5 \times 0.5=$ ) 0.125;
[7] (a) is always expressed(in the phenotype) / produces (functional) proteins;
(b) codominance;

1

4
(d) (i) $\operatorname{sperm}($ with more DNA) have $X$ chromosome;

X is larger / has more genes than Y ;
(ii) female for milk / males for meat / male or female for breeding;
[9] (i) female XX, male XY;

Y shorter / smaller than $X$;
(ii) haemophilia is a recessive allele; defective allele
(gene) present on X , missing from Y ; male $0.5(50 \%$
/ $1 / 2$ ) probability of haemophilia; female $0 /$ no chance;
(0.25(25\% / 1/4) first baby having haemophilia);
or
$X_{H} X_{h} \quad X_{H} Y$;
$X^{H} X^{H}+X^{H} X^{h}+X^{H} Y+X^{h} Y$;
$X^{h} Y$ is a sufferer
(a) mutations;

14 which are different / at different positions in the gene;
(b) (i) either dominant or recessive allele;
(ii) $A^{h} A^{h} B B, A^{h} a B B, A^{h} A^{h} B b, A^{h} a B b$;
(allow 1 mark for 2 or 3 correct answers)
2
(c) if allele A is present (normal) tyrosinase / enzyme is produced, so it doesnot matter what other allele is present / explanation of why heterozygote is same phenotype as double dominant in terms of enzyme produced; phenotype / rabbit is black as both have alleles $A$ and $B$;
one gene influences the expression of another / description using example in question;
(b) aaDD, aa Dd (or DDaa, Ddaa);
(iii) cross with black individual / genotype aaDd or aaDD;genotype is Aadd if agouti offspring / genotype is aadd if no agouti offspring; Accept, repeat
cross using original parents many times; ratio is 4 albino : 3 agouti : 1
black if Aa , or 2 albino : 1 agouti : 1 black if aa;

16 (a) (i) paternal grandmother: $\mathbf{X}^{G} \mathbf{X}^{G}$ or $\mathbf{X}^{G} \mathbf{X}^{\boldsymbol{g}}$

1
(ii) grandparent genotypes: $\left[\mathbf{X}^{\mathbf{g}} \mathbf{Y}\right]\left[\mathbf{X}^{g} \mathbf{X}^{g}\right]\left[\mathbf{X}^{g} \mathbf{Y}\right]$; gametes: $\left[\mathbf{X}^{\mathbf{G}}\right.$ and $\mathbf{X}^{g}$, or $\mathbf{X}^{G}$ only] [ $\mathbf{X}^{\mathbf{g}}$ and $\left.\mathbf{Y}\right]\left[\mathbf{X}^{\mathrm{g}}\right]\left[\mathbf{X}^{\mathrm{g}}\right.$ and $\left.\mathbf{Y}\right]$; parents genotypes: $\left[\mathbf{X}^{\mathrm{G}} \mathbf{Y}\right]\left[\mathbf{X}^{\mathbf{g}} \mathbf{X}^{\mathrm{g}}\right]$ gametes: $\left[\mathbf{X}^{\mathbf{G}}\right.$ and $\left.\mathbf{Y}\right]\left[\mathbf{X}^{\mathbf{g}}\right]$ daughter: $\left[\mathbf{X}^{\mathbf{G}} \mathbf{X}^{\mathbf{g}}\right]$;
(all correct = 3 marks);
(max 2 if no distinction between pairs of gamete genotypes, e.g. comma, space or circle);
(allow omission of gametes clearly not involved in next generation);
(all males $X Y$ and females $X X=1$ mark, if no other marks);
(iii) nil;
$X$ chromosome, without $\mathbf{G}$ allele, inherited from mother / Y must be inherited from father, not $\mathbf{X}^{\mathbf{G}}$;
(b) X and Y chromosomes are different sizes / shapes; chromatids unable to line up and form bivalent / only short pairing region / most of length not homologous;
(i) chromosomes are arranged in (homologous) pairs / bivalents;crossing over / chiasma present / exchange of genetic information; bivalents arranged independently;
(ii) separation / spliting / pulling apart of homologous chromosomes / pairs of chromosomes;
(must give indication that one chromosome moves to each side) (must be in the context of meiosis - not chromatid movements and not chromosomes separate)
pulled at centromere / by spindle / fibres;
(c) (i) the short arm of both chromosomes labelled on the middle homologous pair;
( $\mathbf{B}$ and $\mathbf{b}$ must be labelled on separate chromosomes)
(ii) $8=2$ marks; working showing genotypes with 1 allele from each pair
(for example, B C D) $=1$ mark
(a) gene located on $\mathrm{X} / \mathrm{Y} /$ one sex chromosome;
(allow gene on $X$ or $Y$ chromosome, not $X$ and $Y$ )
(b) (i) black;
(ii) $\quad \mathbf{X}^{\boldsymbol{G}} \mathbf{X}^{g}$;
(lose this mark if the wrong genotype is given for the female in (iii)) (must show $X$ chromosomes to gain the mark)
correct parent gametes
( $\mathbf{X}^{g}$ and $\mathbf{Y}$ from male, $\mathbf{X}^{G}$ and $\mathbf{X}^{g}$ from female);
correct offspring genotypes ( $\mathbf{X}^{\mathbf{g}} \mathbf{X}^{\boldsymbol{g}}, \mathbf{X}^{\boldsymbol{G}} \mathbf{X}^{\boldsymbol{g}}, \mathbf{X}^{\mathbf{G}} \mathbf{Y}, \mathbf{X}^{\mathbf{g}} \mathbf{Y}$ );
correct link of offspring genotypes with phenotypes;
$\mathbf{X}^{9} \mathbf{X}^{g}$ black female
$\mathbf{X}^{G} \mathbf{X}^{g}$ tortoiseshell female
$X^{G} \mathbf{Y}$ ginger male
$\mathbf{X}{ }^{\mathbf{Y}} \mathbf{Y}$ black male
(correct gametes, offspring genotypes and link with phenotypes based on incorrect parent genotype $=3$ marks)
(c) $\mathbf{X}^{\mathbf{G}} \mathbf{Y} \mathbf{d d}$; correct male kitten genotypes ( $\mathbf{X}^{\mathbf{g}} \mathbf{Y} \mathbf{D d}$ and $\mathbf{X}^{\mathbf{g}} \mathbf{Y} \mathbf{d d}$ ); correct link of kitten genotypes with phenotypes;
(ignore female kittens)
$\mathbf{X}^{\mathbf{g}} \mathbf{Y}$ Dd black
$X^{9} Y$ dd grey
(correct kitten genotypes and phenotypes based on incorrect parent genotype = 2 marks)
(a) sandy stated as heterozygous / suitable allusion to alleles;
explained why could not be codominance;
N.B. Second two points linked, not stand alone

| Suitable cross | Reason why not codominance |
| :---: | :---: |
| 3 and 4 | Offspring should all be sandy |
| 10 and 11 | Offspring should all be sandy |
| 7 and 8 | Offspring should all be red |

BUT if candidate assumes sandy is homozygous, mark accordingly e.g. "look at cross 1 and 2; all their offspring would be sandy;" and not that, if red or white then identified as heterozygote, then full 3 marks are still possible.
(b) 11 aabb,
$10=\mathrm{AaBb}$, (N.B. only possibility, not $A-B-)$
2 = A_bb or aa B- (or one possible genotype);
if all 3 correct - 2 marks / if 2 correct - 1 mark; one or fewer - 0 marks
(c) 1 mark for each element of clear explanation i.e.

- choice of a suitable piece of evidence;
- explaining why Hypothesis 2 could not account for the observed result;
(only cross really possible is 1 and 2) i.e. if sandy was aaB_, individuals 1 and 2 would both have been aaB; so their offspring could only be either white or sandy (as no A alleles present);
(d) (Mark line by line, not to 'first error': do not allow for consequential errors)

Individual 18 Other parent
Parental
genotypes AaBb; No mark for
this
Parental gametes AB Ab aB ab and $\mathbf{A b} \mathbf{a b}$;
Offspring
genotypes

| AABb | Aabb | AaBb | Aabb |
| :---: | :---: | :---: | :---: |
| AaBb | Aabb | aaBb | aabb |

(Punnett not necessary
phenotypes red sandy white

## Expected ratio

4
1;
[11] (a) (Gene 1) allele A makes enzyme converting J to $\mathrm{K} /$ colourless to red;

Allele a produces no / non-functional enzyme;
(Gene 2) allele B makes enzyme converting K to $\mathrm{L} /$ red to purple;
Allele b produces no / non-functional enzyme;
("Recessive alleles produce no / non-functional enzyme" = 2)
White flowers result from genotype aa;
... regardless if $B$ or $b /$ even if aaB_;
Colourless (substance) / J produces white;
Red flowers when A_bb / enzyme 1 only;
Purple flowers when A_B_/ enzymes 1 and 2;
6 max
(b) (i) (1) (red parent) AAbb;
(2) (white parent) aaBB;
(ii) $\mathrm{F}_{1}$ are AaBb ;
$F_{2}$ ratio of $9: 3: 4$;
Purple : red : white;
Suitable working shown;
(c) (i) aabb, aaBb, and aaBB; (allow aabb \& aaB_)
(ii) (Crush each type of white petal to make an extract, and) add some of the (red) pigment / K, to petal OR incubate with K;
(extract becoming) purple is identified as aaBB OR that staying red, after K is added, is aabb;
[15] (a) Correct answer: 1.25;

## Ignore working

OR (if wrong answer)
$\frac{\text { measurement in } \mu \mathrm{m}}{40000}, \frac{\text { measurement in } \mathrm{mm}}{40}=1$ mark
125 but wrong order of magnitude $=1$ mark
2 (ii) C has myosin / thick (and actin / thin) filaments;

OR
A has only actin / thin (/ no myosin / no thick) filaments;
(b) When contracted:

Thick \& thin filaments/myosin \& actin overlap more;
Interaction between myosin heads \& actin / cross-links form;
Movement of myosin head;
Thin filaments / actin moved along thick filaments / myosin;
Movement of thin filaments / actin pulls Z-lines closer together;
Displacement of tropomyosin to allow interaction;
Role of Ca ;
Role of ATP;
Allow ref. to 'sliding filament mechanism'/ described if no other marks awarded
(c) (i) 8 has DMD but 3 and 4 do not / 12 has DMD but 6 and 7 do not / neither parent has the condition but their child has;

Allow parents 3 and 4 give 8, parents 6 and 7 give 12
(ii) 4 AND 7;
(iii) Parental genotypes: $6=\mathbf{X}^{\mathrm{D}} \mathbf{Y}$ AND 7 $=\mathbf{X}^{\mathrm{D}} \mathbf{X}^{\mathrm{d}}$

AND
Gametes correct for candidate's P genotypes - e.g.
$\mathbf{X}^{\mathrm{D}}$ and $\mathbf{Y}+\mathbf{X}^{\mathrm{D}}$ and $\mathbf{X}^{\mathrm{d}}$;
Offspring genotypes correctly derived from gametes e.g.
$\mathbf{X}^{\mathrm{D}} \mathbf{X}^{\mathrm{D}}+\mathbf{X}^{\mathrm{D}} \mathbf{X}^{\mathrm{d}}+\mathbf{X}^{\mathrm{D}} \mathbf{Y}+\mathbf{X}^{\mathrm{d}} \mathbf{Y} ;$

Male offspring with MD correctly identified: $\mathbf{X}^{\mathrm{d}} \mathbf{Y}$;
Probability $=0.25$ / correct for candidates offsprings genotypes;
(d) (i) No gene fragment $\mathbf{G}$;
(ii) Only one copy of gene fragment $\mathbf{F}$;

Male has only one X -chromosome / is XY (c.f. female has two / is XX );

2

3
(e) (i) To prevent rejection / prevent antibody production vs. injected cells / injected cells have (foreign) antigen (on surface);
(ii) Shows effect of cells / not just effect of injection / not just effect of salt solution;
(iii) Only one person tested so far - need more to see if similar results /need more to see if reliable;

Need to assess if new (dystrophin positive) muscle fibres are functional / if muscle becomes functional;

Can't tell how widespread effect is in the muscle / sample taken near injection site;

Need to test for harmful side effects;
Need to test if successful for other mutations of dystrophin gene;
Need to assess permanence / longevity of result/insufficient time allowed in investigation;
(In this patient) only small response / \%;
Further sensible suggestion;

## 4 max

[25] (a) Parents without CF $\rightarrow$ offspring with CF / 1 + $2 \rightarrow 6 / 7+8 \rightarrow 10$;

Each parent must have CF allele / offspring receives CF allele from both parents / both parents heterozygous / both carriers;
(b) $\quad \mathbf{N n}$ and $\mathbf{N n}$ (no mark since awarded in (a) already)

Accept alternative symbols
$\mathbf{N} \quad \mathbf{n}$ and $\mathbf{N} \quad \mathbf{n}$;
Ignore $X$ and $Y$
$\mathbf{N N}$ and $\mathbf{N n}$ and $\mathbf{N n}$ and $\mathbf{n n}$;
Correct allocation of phenotypes to genotypes;
Probability $=\underline{0.125 ;}$
Accept answers expressed as chance rather than probability, eg 1 in 8 / 1 to 7 / 12.5\%;
[6] (a) Daughter (C) does not have the condition / one child doesn't have it;

Accept converse arguments (If candidates see it purely as a genetic cross diagram) $D$ is heterozygous because $E$ is unaffected;

Parents must have been carriers of normal / healthy recessive/ if recessive then parents homozygous (so all children affected);
$D$ has cancer, so the cancer allele must be dominant;
(b) Father (A) would pass on X chromosome to daughter; She is not affected;

Accept that if D's X chromosome carried 'it', then $E$ would be affected.
(c) Only 25 / young so don't know if cancer will develop;

Accept E must be homozygous recessive/have two recessive alleles;

Don't know if her father was heterozygous or homozygous;
So no chance of cancer / no more chance than rest of the population;

If heterozygous, she has a $50 \%$ chance of carrying the allele/gene; If homozygous, she has a serious risk of cancer.
(d) Mutation / mutagen changes DNA of cell;

Damaged DNA not repaired / cells not killed / apoptosis doesn't happen; Mutation leads to loss of control / uncontrolled cell division; (Some of these) cells carried to other parts of the body.

## 3 max

