### Mark schemes

1

Parental genotypes:	Gg nn	gg Nn	•	
Gamete genotypes	Gn gn	gN	gn	;

	gN	gn
Gn	Gg Nn	Gg nn
	Grey, normal	Grey, vestigial
gn	gg Nn Ebony, normal	gg nn Ebony, vestigial

All offspring genotypes correct;

All offspring genotypes correctly derived;

[4] (a) (i) Only seen in males / not in females;

2		1	
	(ii) Unaffected parents / mother $\rightarrow$ child with M.D. / (1 ×)2 $\rightarrow$ 5 / (3 ×) 4 $\rightarrow$ 11 / 8 (× 9) $\rightarrow$ 13;	1 (b) 5 = X <sup>d</sup> Y	
	$6 = X^{D}Y$		
	$7 = X^{D}X^{d} \underline{AND} X^{D}X^{D}$		
	$8 = X^{D}X^{d};;$		
	All 4 correct = 2 marks		
	2 or 3 correct = 1 mark		
		max 2	
(c)	¼ / 0.25 / 25% / 1:3 / 1 in 4; ( <i>NOT</i> '1:4')		
		1	.,
			[:
] (a	) Cannot make (active) enzyme A (which converts precursor to linamar	rin) / cannot make	

l n a m a

#### (b) (i) **AL** + **AI** + **aL** + **aI** ;

 Meiosis separates alleles / homologous chromosomes / pairs of chromosomes; Independent assortment / means either of A / a can go with either of L / I; [Accept: 'random segregation'] [Cancel: if reference to crossing-

over]

2

r i n

1

1

# (d) From parental genotypes: AaLI × 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	AaLl
AI	AALI	AAII	AaLl	Aall
aL	AaLL	AaLl	aaLL	aaLl
al	AaLl	Aall	aaLl	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%;

- (e) (i) There was no (significant) difference in damage between cyanogenic andacyanogenic / being cyanogenic has no effect;
  - (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 <u>only</u> cyanogenic plants / only cyanogenic plants survive;
  - 2. (Cyanide release) limits / stops feeding by slugs / slugs killed;

3

1

;

Low slug population:

4

3. Find both types of plant;

 Less selection pressure from slugs / no selective advantage / no selection /described;

4 [15] (a) Gg / suitable equivalent;

Grey : black about 3: 1; [Note: Can be in table / diagram]

- (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)  $P^2 = (0.25)^2 / 0.0625 / \text{square of calculated figure for } C^N; = 2 \text{ marks } p^2 + 2pq + q^2 = 1.0; = 1 \text{ mark}$ = 31.25 / 31; [Accept: Derived from either  $p^2$  or  $q^2$ ]
- 3

1

- [10]
- (a) Only expressed in the homozygote / not expressed in the heterozygote / not expressed if

**5** dominant present;

(b) Tt Aa × tt aa ;

ta

TA Ta tA

	ТА	Та	tA	ta	
ta	TtAa	Ttaa	ttAa	ttaa	;
	Orange striped	Orange unstriped	White striped	White unstriped / snowy	;

ta ;

2

2

1

	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 <u>MUST</u> 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					
		<b>[7]</b> (a) (i)	$2 \max$ BBX <sup>A</sup> Y BbX <sup>A</sup> Y				
6			1				
		(ii) BbX <sup>A</sup> X <sup>a</sup> , bbX <sup>A</sup> X <sup>a</sup> ;	1				
	(b)	parental genotypes – BbX <sup>A</sup> Y x BbX <sup>A</sup> X <sup>a</sup> ;	1				
		<i>Gametes</i> – $(BX^A, bX^A,) BY, bY, BX^A, BX^a, bX^A, bX^a;$	1				
		Genotypes of sons- ;					

		Male gametes	
		BY	bY
	BX <sup>A</sup>	BBX <sup>A</sup> Y	BbX <sup>A</sup> Y
Female	B X <sup>a</sup>	BB Xª Y	Bb Xª Y
gametes	bX <sup>A</sup>	BbX <sup>A</sup> Y	bbX <sup>A</sup> Y
	b X <sup>a</sup>	Bb Xª Y	bb Xª Y

1 [6] (a) aabb;

0.125 / 12.5% / 1/8 ;

7

(b) AaBb and aabb;

(c) Pea comb offspring will produce blue eggs;

	Alleles <b>A</b> and <b>B</b> are inherited together / are on the same chromosome;	2
(d)	Reference to crossing over; Reduce chance of genes being separated (by crossing over); If crossing over occurred some gametes will contain alleles <b>A</b> and <b>b</b> ;	2 max
(e)	Two suitable environmental factors;	
	e.g. Diet / named component of diet; Temperature; Light intensity / duration; Disease;	2 max
(f)	Cross C / X <sup>f</sup> X <sup>f</sup> and X <sup>F</sup> 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;	2
(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
		<i>D</i> , 111 <i>Da</i> ,
	(both correct 1 mark)	1
(b)	Epistasis; One gene controlling / inhibiting the expression of another;	2
(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	
	Ratio 2 1 1 ;	3 [6]

(a) Two linked points: Crossing over / exchange of material (between chromatids); Different combinations of alleles / linkage groups changed / broken; OR Independent assortment / alignment of (homologous) chromosomes; Different combinations of (maternal and paternal) chromosomes / alleles; 2 max (b) D d М Gamete genotype m; D d M Offspring genotype m; Offspring Abnormal males / (all) (no females); phenotypes 3 [5] (a) black; (i) 10 (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] (a) males are XY and females XX / 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 - hh X <sup>b</sup> Y; 6 - Hh X <sup>B</sup> X <sup>b</sup> ;	
			2
	(ii)	h X <sup>b</sup> , h Y, and H X <sup>B</sup> , h X <sup>B</sup> , H X <sup>b</sup> , hX <sup>b</sup> ;	
			1
	(iii)	1 / 8 or 12.5% or 0.125;;	
		either genetic diagram to show genotypes Hh $X^b$ $X^b$ , Hh $X^BY$ , hh	
		$A^{-} A^{-}, \Pi \Pi A^{-} T, \Pi \Pi A^{-} A^{-}, \Pi \Pi A^{-} T, \Pi \Pi A^{-} A^{-}, \Pi \Pi A^{-} T,$ 1 / 8:	
		or	
		P (boy) = 0.5, P (colour blind) = 0.5, P (white streak) = $0.5$ ;	
		$(0.5 \times 0.5 \times 0.5 =) 0.125;$	
			2
		[7] (a) is always expressed(in the phenotype) / produces (functional)	proteins;

2 max

12			1
	(b)	codominance;	1
	(c)	Parental geneotypes - $hhCC$ , $HhCC$ ;	
		Gametes-	
		Offspring geneotypes - HhCC, hhCC, HhCC, hhCC; Offspring pheneotypes - hornless horned hornless horned	
	roan	roan white white	
		Ratio of offspring - 1 1 1 1;	4
	(d)	<ul><li>(i) sperm(with more DNA) have X chromosome;</li><li>X is larger / has more genes than Y;</li></ul>	
			2
		(ii) female for milk / males for meat / male or female for breeding;	
		<b>[9]</b> (i)	1 female XX, male XY
13			
_		Y <u>shorter / smaller</u> than X;	2
	(ii)	haemophilia is a recessive allele; defective allele	

(gene) present on X, missing from Y; male 0.5(50%

		/ ½) probability c chance; (0.25(25% / ¼) f	f haemophilia; female 0 / no rst baby having haemophilia);		
		or			
		ХнХh ХнY; Х <sup>н</sup> Х <sup>н</sup> + Х <sup>н</sup> Х <sup>h</sup>	+ X <sup>H</sup> Y + X <sup>h</sup> Y;		
		X <sup>h</sup> Y is a suffere	r		
				3 max	[5]
	(a)	mutations;			
<b>14</b> v	vhich	re different / at d	ifferent positions in the gene;	2	
	(b)	(i) either dom	inant or recessive allele;	1	
		(ii) A <sup>h</sup> A <sup>h</sup> BB, A	<sup>h</sup> aBB, A <sup>h</sup> A <sup>h</sup> Bb, A <sup>h</sup> aBb;		
		(allo	w 1 mark for 2 or 3 correct answers)	2	
		(iii) temperatur denatured;	e lower at extremities;enzyme active / not		
	(c)	if allele A is pres what other allele as double domir both have alleles	ent (normal) tyrosinase / enzyme is produced, so it doesnot matter is present / explanation of why heterozygote is same phenotype ant in terms of enzyme produced; phenotype / rabbit is black as s A and B; [9] (	2 a) epist	asis;
15		one gene influer using example ir	ces the expression of another / description a question;	2	
	(b)	aaDD, aa Dd (or	DDaa, Ddaa);	1	
	(c)	(i) AaDd (or I	DdAa);	1	
		(ii) aadd, Aad	d (or ddaa, ddAa);	1	
		(iii) cross with agouti offs	black individual / genotype aaDd or aaDD;genotype is Aadd if pring / genotype is aadd if no agouti offspring; <i>Accept</i> ; 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;	2	[7]
<b>16</b> (a)	(i	)	paternal grandmother: X <sup>G</sup> X <sup>G</sup> or X <sup>G</sup> X <sup>g</sup>		[,]
				1	
		(ii)	grandparent genotypes: [X <sup>g</sup> Y] [X <sup>g</sup> X <sup>g</sup> ] [X <sup>g</sup> Y]; gametes: [X <sup>G</sup> and X <sup>g</sup> , or X <sup>G</sup> only] [X <sup>g</sup> and Y] [X <sup>g</sup> ] [X <sup>g</sup> and Y]; parents genotypes: [X <sup>G</sup> Y] [X <sup>g</sup> X <sup>g</sup> ] gametes: [X <sup>G</sup> and Y] [X <sup>g</sup> ] daughter: [X <sup>G</sup> X <sup>g</sup> ]; (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 XY and females XX = 1 mark, if no other marks);	3	
		(iii)	nil; X chromosome, without <b>G</b> allele, inherited from mother / Y must be inherited from father, not <b>X</b> <sup>G</sup> ;	2	
(	b)	X ar chro sho	nd Y chromosomes are different sizes / shapes; omatids unable to line up and form bivalent / only rt pairing region / most of length not homologous;	2	
				2 [8] (a)	6;
17				1	
		(i)	chromosomes are arranged in (homologous) pairs / bivalents;crossing over / chiasma present / exchange of genetic information; bivalents arranged independently;	2 max	
		(ii)	separation / spliting / pulling apart of <u>homologous</u> chromosomes / <u>pairs</u> 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;	2	
(	c)	(i)	the short arm of both chromosomes labelled on the middle homologous pair;		
			( <b>B</b> and <b>b</b> must be labelled on separate chromosomes)	1	

		<ul> <li>8 = 2 marks; working showing genotypes with 1 allele</li> <li>from each pair</li> </ul>		
		(for example, <b>B C D</b> ) = 1 mark	2	
			2	[8]
				[•]
	(a)	gene located on X / Y / one sex chromosome;		
18				
		(allow gene on X or Y chromosome, not X and Y)	1	
			-	
	(b)	(i) black;	1	
			1	
		(ii) X <sub>G</sub> X <sub>a</sub> ;		
		(lose this mark if the wrong genotype is given for the female in (iii))		
		(must show X chromosomes to gain the mark)	1	
			1	
		correct parent gametes		
		(X <sup>y</sup> and Y from male, X <sup>y</sup> and X <sup>y</sup> from female); correct offspring genotypes (X9X9, X <sup>G</sup> X9, X <sup>G</sup> Y, X9Y).		
		correct link of offspring genotypes with phenotypes;		
		X <sup>g</sup> X <sup>g</sup> black female		
		X <sup>G</sup> X <sup>g</sup> tortoiseshell female		
		X <sup>G</sup> Y ginger male		
		X <sup>g</sup> Y black male		
		(correct gametes, offspring genotypes and link with phenotypes		
		based on incorrect parent genotype = 3 marks)	3	
			5	
	(c)	X <sup>G</sup> Y dd; correct male kitten genotypes (X <sup>g</sup> Y Dd and		
		X <sup>g</sup> Y dd); correct link of kitten genotypes with		
		phenotypes;		
		(ignore temale kittens)		
		X <sup>g</sup> Y Dd black		
		X <sup>g</sup> Y dd grey		
		(correct kitten genotypes and phenotypes based on incorrect parent		
		genotype = 2 marks)	2	
			3	[9]
10	(a)	sandy stated as neterozygous / suitable allusion to alleles;		

suitable cross chosen; (as in table)

explained why could not be codominance;

N.B. Second two points linked, not stand alone

Suitable cross	Reason why <b>not</b> 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 = 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 <i>this</i>	AaBb;	No mark for (AaBb)
Parental gametes AB Ab aB al	b and	Ab ab;

Parental gametes AB Ab aB ab

Offspring

genotypes

AABb	Aabb	AaBb	Aabb
AaBb	Aabb	aaBb	aabb
(Dunnatt not nagagagan)			

<sup>(</sup>Punnett not necessary

3

2

	Offs phei	pring notypes	red	sandy	white		
	Exp	ected ratio	3	4	1;		
			[11] (a)	(Gene 1) allele A	makes enzyme conve	erting J	4 to K / colourless to red;
	Allel (Ger Allel ("Re Whit re Colo Red Purp	e a produces ne 2) allele E e b produces cessive allel e flowers res gardless if E ourless (subs flowers whe ole flowers w	s no / non-fi b makes <u>enz</u> s no / non-fi es produce sult from ge s or b / even stance) / J p n A_ bb / ei hen A_ B_ ,	unctional <u>enzyme</u> <u>zyme</u> converting unctional <u>enzyme</u> no / non-functior motype aa; n if aaB_ ; roduces white; nzyme 1 only; / enzymes 1 and	2; K to L / red to purple; 2; nal enzyme" = 2) 2;		6 max
(b)	(i)	(1) (red pa	rent) AAbb;				
		(2) (white p	oarent) aaB	В;			2
	(ii)	$F_1$ are AaB $F_2$ ratio of $F_2$ Purple : re Suitable w	b; 9 : 3 : 4; d : white; orking show	vn;			4
(c)	(i)	aabb, aaBl	b, and aaBB	3; (allow aabb & a	aaB_)		1
	(ii)	(Crush eac pigment / k (extract be added, is a	h type of wl (, to petal O coming) pu aabb;	hite petal to make R incubate with I rple is identified a	e an extract, and) add s K; as aaBB OR that stayir [1	some o ng red, <b>5]</b> (a)	f the (red) after K is 2 Correct answer: 1.25;
		Ignoi	re working				
	OR	(if wrong a	nswer)				
	mea	asurement in 40000	uµm mea	surement in mm 40	= 1 mark		
		125	but wrong o	rder of magnitud	e = 1 mark		

2 (ii) **C** has myosin / thick (and actin / thin) filaments;

		A has only actin / thin (/ no myosin / no thick) filaments;	1 max				
(b)	Whe	n contracted:					
	Thick	& thin filaments/myosin & actin overlap more;					
	Intera	action 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;						
	Displ	acement of tropomyosin to allow interaction;					
	Role of Ca ;						
	Role of ATP;						
		Allow ref. to 'sliding filament mechanism' / described if no other marks awarded	4 max				
(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	1				
	(ii)	4 <b>AND</b> 7;	1				
	(iii)	Parental genotypes: $6 = \mathbf{X}^{D}\mathbf{Y}$ AND $7 = \mathbf{X}^{D}\mathbf{X}^{d}$					
		AND					
		Gametes correct for candidate's P genotypes – e.g.					
		$\mathbf{X}^{D}$ and $\mathbf{Y} + \mathbf{X}^{D}$ and $\mathbf{X}^{d}$ ;					

Offspring genotypes correctly derived from gametes e.g.

 $\mathbf{X}^{\mathsf{D}}\mathbf{X}^{\mathsf{D}} + \mathbf{X}^{\mathsf{D}}\mathbf{X}^{\mathsf{d}} + \mathbf{X}^{\mathsf{D}}\mathbf{Y} + \mathbf{X}^{\mathsf{d}}\mathbf{Y};$ 

Male offspring with MD correctly identified:  $\boldsymbol{X}^{d}\boldsymbol{Y};$ 

Probability = 0.25 / correct for candidates offsprings genotypes; Accept  $\frac{1}{4}$  / 1 in 4 / 1:3 / 25%

			4
(d)	(i)	No gene fragment <b>G</b> ;	1
	(ii)	Only one copy of gene fragment <b>F</b> ;	
		Male has only one X-chromosome / is XY (c.f. female has two / is XX);	2
	(iii)	10 has only one copy of gene fragment <b>G</b> ;	
		10 has only one normal X-chromosome / has one abnormal / dDdhas only one normal allele / has one X / is X X / is heterozygous;	
		11 has two normal X-chromosomes / has 2 normal alleles / DD d is X X / has not got X / has 2 copies of (F and) G;	3
(e)	(i)	To prevent rejection / prevent antibody production vs. injected cells / injected cells have (foreign) antigen (on surface);	1
	(ii)	Shows effect of <u>cells</u> / not just effect of injection / not just effect of salt solution;	1
	(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  $\rightarrow$  I0;

from both parents / both parents heterozygous / both carriers; 2 (b) **Nn** and **Nn** (no mark since awarded in (a) already) Accept alternative symbols Ν **n** and **N n**; Ignore X and Y NN and Nn and Nn and nn; Correct allocation of phenotypes to genotypes; Probability = 0.125; Accept answers expressed as chance rather than probability, eg 1 in 8 / 1 to 7 / 12.5%; 4 Daughter (C) does not have the condition / one child doesn't have it; [6] (a) 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; 2 (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. 2 (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;

Each parent must have CF allele / offspring receives CF allele

22

23

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