Chapter 10 Genetics and Evolution *[172 marks]*

1. In which situation are alleles exchanged? *[1 mark]*

A. During the separation of sister chromatids
B. In the transmission of linked genes
C. During fertilization when sperm and egg chromosomes pair up
D. When chiasmata are formed between non-sister chromatids

Markscheme

D

2. The statement is about the genetic control of cat coat colour. *[1 mark]*

> Many genes control cat coat colour. Tabby cats have a coat with stripes. Two of their genes are the tabby gene Mm and the agouti gene Aa.
> - The dominant allele **M** codes for straight stripes, whereas the recessive allele **m** codes for blotches and whorls instead of stripes.
> - The dominant allele **A** produces two-toned hair, to reveal coat patterns, whereas the recessive allele **a** causes all coat hair to be black and so hides any pattern.

In a cross between two double heterozygous tabby cats, what would the expected proportion of black offspring be?

A. 1 out of 16
B. 3 out of 16
C. 4 out of 16
D. 9 out of 16

Markscheme

C

3. This is the cross that led to the discovery of non-Mendelian ratios in Morgan's experiments with *Drosophila*. *[1 mark]*

Grey body – Normal wings $\frac{b^+ \quad vg^+}{b \quad vg}$ × Black body – Vestigial wings $\frac{b \quad vg}{b \quad vg}$

Which is a recombinant genotype?

A. $\frac{b^+ \quad vg^+}{b \quad vg}$

B. $\frac{b \quad vg}{b \quad vg^+}$

C. $\frac{b^+ \quad vg^+}{b^+ \quad vg^+}$

D. $\frac{b \quad vg}{b \quad vg}$

Markscheme

B

4. During which stage of meiosis does crossing over usually occur? *[1 mark]*

A. Prophase I

B. Metaphase I

C. Prophase II

D. Metaphase II

Markscheme

A

5. At which stage of meiosis does a pair of sister chromatids separate? *[1 mark]*

A. Metaphase I

B. Anaphase I

C. Metaphase II

D. Anaphase II

Markscheme

D

6. In a fruit fly experiment, grey body, normal winged (homozygous dominant) fruit flies were mated with black body, short winged (homozygous recessive) fruit flies. The F_1 dihybrid females were then used in a test cross. If the genes are always linked and no crossing over occurs, what would be the predicted ratio in the F_2 generation? *[1 mark]*

A. 9 : 3 : 3 : 1

B. 1 : 1 : 1 : 1

C. 3 : 1

D. 1 : 1

Markscheme

D

7. Skin colour is a trait controlled by polygenic inheritance. Which statement is correct? *[1 mark]*

A. Skin colour shows discontinuous variation

B. Individuals show a wide range of phenotypes for skin colour

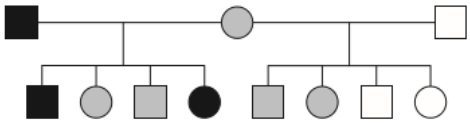
C. No two people have the same skin colour

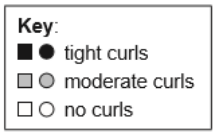
D. Children always have the same skin colour as one of their parents

Markscheme

B

8. The curly hair of the coat of Selkirk Rex cats is due to the presence of the allele SC. These cats can either have tight curls or be moderately curly, whereas the coat of other cats is usually made of straight hair with no curls because of the allele SS. Circles indicate female cats and squares indicate males. *[1 mark]*





What are the phenotypes of cats with these genotypes?

	$S^S S^S$	$S^S S^C$
A.	no curls	moderate curls
B.	tight curls	no curls
C.	tight curls	moderate curls
D.	no curls	tight curls

Markscheme

A

9. Which is a statement of Mendel's law of independent assortment? *[1 mark]*

A. Allele pairs separate during gamete formation and recombine during fertilization.
B. Allele pairs for different genes separate independently during gamete formation.
C. Unlinked alleles are assorted with a 9 : 3 : 3 : 1 ratio in a dihybrid cross.
D. Allele pairs for the same gene are assorted independently during gamete formation.

Markscheme

B

10. In a variety of tulips, V is the allele for variegated colour and C is the allele for compound flower. Which cross will give a 1:1:1:1 ratio of phenotypes in the offspring? *[1 mark]*

A. VvCc × VvCc
B. VVcc × vvCC
C. VvCc × vvCc
D. Vvcc × vvCc

Markscheme

D

11. In fruit flies (*Drosophila melanogaster*) grey body is dominant to black body and long wings are dominant to vestigial wings. Two flies heterozygous for both genes were crossed. What proportion of the offspring would be expected to have black bodies and long wings? *[1 mark]*

A. 1/2
B. 3/16
C. 1/4
D. 1/16

Markscheme

B

12. Human skin colour shows continuous variation. What does this indicate about the pattern of inheritance of human skin colour? *[1 mark]*

A. It is dominant.
B. It is sex-linked.
C. It is recessive.
D. It is polygenic.

Markscheme

D

13. How does meiosis cause Mendel's law of independent assortment? *[1 mark]*

A. Linked genes are randomly separated.
B. The chromosome number is divided twice.
C. Crossing-over occurs in Anaphase I.
D. Alleles that are not in the same linkage group are segregated.

Markscheme

D

14. A test cross of **linked** genes was performed with fruit flies (*Drosophila melanogaster*). *[1 mark]*

Wild type body (B) is dominant to black body (b)
Normal wings (W) is dominant to vestigial wings (w)

BbWw crossed with bbww

The resulting offspring were	952 wild type body, normal wings
	948 black body, vestigial wings
	200 wild type body, vestigial wings
	198 black body, normal wings

What is the most likely explanation for these results not fitting the expected ratio?

A. Crossing-over
B. Non-disjunction
C. Gene mutation
D. Random variation

Markscheme

A

15. Why do humans inherit continuous variation with regard to height? *[1 mark]*

A. The trait for tallness is dominant.
B. The height phenotype is polygenic.
C. This is a case of multiple alleles.
D. Height in humans is polyclonal with multiple alleles.

Markscheme

B

16. Maize (*Zea mays*) contains 20 chromosomes in a diploid cell. How many chromosomes will be in each cell after the first and second division of meiosis? *[1 mark]*

	After first meiotic division	After second meiotic division
A.	10	10
B.	20	10
C.	40	20
D.	10	5

Markscheme

A

17. What causes variation in **both** sexually and asexually reproducing organisms? *[1 mark]*

A. Mutations
B. Polygenic inheritance
C. Crossing over
D. Independent assortment

Markscheme

A

18. What causes genetic variety in the formation of gametes during meiosis? *[1 mark]*

A. Crossing over in prophase I and random orientation of homologous chromosomes in metaphase I
B. Crossing over in metaphase I and random orientation of homologous chromosomes in metaphase II
C. Linkage of genes in prophase I and crossing over in metaphase I
D. Linkage of genes in metaphase I and random orientation of homologous chromosomes in metaphase II

Markscheme

A

19. When does an unequal division of cytoplasm occur? *[1 mark]*

A. During meiosis in the apical meristem
B. During the division of Sertoli cells into spermatozoa
C. During binary fission of eukaryotic cells
D. During meiosis in the human ovary

Markscheme

D

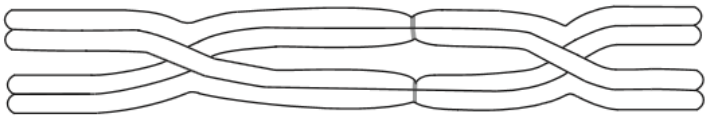
20. What happens in crossing over? *[1 mark]*

A. Exchange of genetic material between homologous chromosomes
B. Exchange of genes during metaphase of mitosis
C. Random distribution of chromosomes during meiosis
D. Homologous chromosomes fail to separate during meiosis

Markscheme

A

21. The diagram below shows chromosomes during meiosis. *[1 mark]*



How many chromosomes and chiasmata are visible?

	Chromosomes	Chiasmata
A.	4	4
B.	2	4
C.	2	2
D.	4	2

Markscheme

C

22. What is a suspected heterozygous individual crossed with in a test cross? *[1 mark]*

A. Homozygous dominant
B. Homozygous recessive
C. Heterozygous dominant
D. Heterozygous recessive

Markscheme

B

23. A test cross resulted in these recombinants: *[1 mark]*

$$\frac{tB}{tb} \qquad \frac{Tb}{tb}$$

Which of the following was the parental test cross?

A. $\frac{TB}{tb} \times \frac{tb}{tb}$

B. $\frac{TB}{tB} \times \frac{tb}{Tb}$

C. $\frac{Tb}{tB} \times \frac{tb}{tb}$

D. $\frac{TB}{tb} \times \frac{TB}{tb}$

Markscheme

A

24. Which are the possible recombinants in a dihybrid test cross involving the linked genes JQ/jq? *[1 mark]*

A. JQ/jq and JJ/Qq

B. Jq/Qq and Qq/JJ

C. Jq/jq and jQ/jq

D. JQ/jq and Jq/jQ

Markscheme

C

25. How many autosomes are there in a human sperm? *[1 mark]*

A. 1
B. 22
C. 23
D. 46

Markscheme

B

26. What is polygenic inheritance? *[1 mark]*

A. A character that is controlled by two or more genes
B. A character that is controlled by more than two copies of a gene
C. Inheriting more than two alleles of a gene
D. Inheriting a linked group of genes

Markscheme

A

27. When do chiasmata form in meiosis? *[1 mark]*

A. During prophase I
B. During metaphase I
C. During anaphase I
D. During prophase II

Markscheme

A

28. Which of the following processes result in the production of recombinants? *[1 mark]*

I. Crossing over between linked genes
II. Reassortment of non-linked genes
III. Mutation

A. I only
B. I and II only
C. I and III only
D. I, II and III

Markscheme

B

29. Which of the following conclusions did Mendel make from his experiments? *[1 mark]*

A. Dominant genes are more frequent than recessive genes.
B. Genes are composed of DNA.
C. Traits are inherited in discrete units, one from each parent.
D. Segregation occurs through meiosis.

Markscheme

C

30. *[1 mark]*

In fruit flies (*Drosophila melanogaster*) grey body is dominant to black body and normal wings are dominant to vestigial wings.

If a heterozygous grey fruit fly is mated with a black-bodied fruit fly, what proportion of the offspring would be black?

A. 0 %

B. 25 %

C. 50 %

D. 100 %

Markscheme

C

31. *[1 mark]*

In fruit flies (*Drosophila melanogaster*) grey body is dominant to black body and normal wings are dominant to vestigial wings.

Male flies, heterozygous for both grey body and normal wings, were mated with black-bodied, vestigial-winged females. 2000 offspring were counted. The resulting percentage of each type of offspring is shown in the table below.

Resulting offspring	Frequency
Grey body, normal wings	40%
Black body, vestigial wings	40%
Grey body, vestigial wings	10%
Black body, normal wings	10%

What conclusion can be drawn from the information given above?

A. The genes assort independently.
B. A mistake has been made.
C. The genes are linked.
D. The genes are on separate chromosomes.

Markscheme

C

32a. Determine the phenotype of Aabb. *[1 mark]*

Markscheme

tall and yellow

32b. Compare the information that could be deduced when the genotypes are presented as AaBb or $\frac{A\ B}{a\ b}$. *[2 marks]*

Markscheme

both indicate the same phenotypes / both represent tall green plants;

(AaBb indicates unlinked genes and) $\frac{A\ B}{a\ b}$ linked genes;

AaBb indicates genes on different chromosomes and $\frac{A\ B}{a\ b}$ on the same chromosome;

independent assortment/AB, Ab, aB and ab (gametes) with AaBb but not independent assortment/only AB and ab (gametes) with $\frac{A\ B}{a\ b}$ (unless there is crossing over);

32c. Deduce **one** possible recombinant offspring of individual $\frac{A\ B}{a\ b}$ after a test cross. *[1 mark]*

Markscheme

Aabb **or** aaBb **or** $\frac{\text{A b}}{\text{a b}}$ **or** $\frac{\text{a B}}{\text{a b}}$ **or** tall yellow **or** short green

33a. Outline the genetic evidence that *Canis* populations have hybridized. *[2 marks]*

Markscheme

a. (all three) *Canis* populations show a mixture of haplotypes from two (or more) origins;
b. Minnesota-northwestern wolves have a mixture of haplotypes from grey wolf/ *C. lupus* and eastern wolf/ *C. lycaon*;
c. southern Ontario coyote has mixture of haplotypes from western coyote/ *C. latrans* and eastern wolf;
d. northeastern coyote has mixture of haplotypes from western coyote/ *C. latrans* and eastern wolf;

33b. Compare the genetic data for southern Ontario coyotes and northeastern coyotes. *[2 marks]*

Markscheme

a. both contain haplotypes C1, C9 and C19;
b. C19 haplotype in greater proportion in northeastern coyotes / vice versa;
c. C9 haplotype in greater proportion in southern Ontario coyotes / vice versa;
d. C1 in smaller proportion in southern Ontario than northeastern coyotes / vice versa;
e. southern Ontario coyotes / northeastern coyotes have more haplotypes from coyotes than wolves;
f. fewer haplotypes from other sources in northeastern coyotes / vice versa;

33c. State with a reason whether the genetic evidence shows that the western coyote and the grey wolf have overlapping ranges. *[1 mark]*

Markscheme

no overlapping ranges since no haplotypes/C9, C19 from western coyotes present with haplotype/C22 from grey wolf in any of the hybrids.

33d. The northeastern coyote has more wolf-like skull features than the southern Ontario coyote. Suggest a reason for this difference. *[1 mark]*

Markscheme

northeastern coyote has greater proportion of C1/ haplotype from (eastern) wolf (than southern Ontario coyote)

33e. Discuss briefly whether there is genetic evidence to show a common ancestor for the Minnesota-northwestern wolf, the southern Ontario coyote and the northeastern coyote. *[2 marks]*

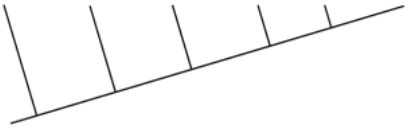
Markscheme

a. all three show evidence that the eastern wolf was their ancestor/all have haplotypes from the eastern wolf;
b. southern Ontario and northeastern coyotes have different eastern wolf haplotypes from the Minnesota northwestern wolf / southern Ontario and northeastern coyotes have C1 while the Minnesota northwestern wolf has C3 and C13;

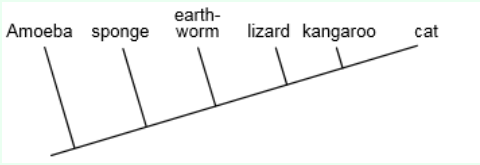
34a. The table shows certain characteristics present (+) or absent (–) in six organisms. *[3 marks]*

	Segmented	Jaws	Hair	Placenta	Multicellular	Limbs
Amoeba	–	–	–	–	–	–
Cat	+	+	+	+	+	+
Earthworm	+	–	–	–	+	–
Kangaroo	+	+	+	–	+	+
Lizard	+	+	–	–	+	+
Sponge	–	–	–	–	+	–

Using the data, label the cladogram with the names of the organisms.



Markscheme



*Award **[1]** for the correct position of any two organisms.*
*Award **[1 max]** if the correct order is reversed horizontally (ie from cat on the left to Amoeba on the right).*

34b. A species is often defined as a group of similar individuals that interbreed in nature and produce fertile offspring. Discuss some problems with the use of this definition. *[2 marks]*

Markscheme

a. not all organisms can be defined in this way / does not take into account hybrids/ microorganisms/plants;
b. (even if able to interbreed) may have differences in DNA/protein;
c. does not apply to bacteria/other organisms that reproduce asexually;
d. in sympatric/allopatric isolation members of the same species do not interbreed;
e. (in some species) significant differences in morphology can occur within the same species eg: sexual dimorphism/metamorphosis/ring species;
Accept any other correct answer.

35a. Analyse this karyotype. *[2 marks]*

Markscheme

Male has (one X and) one Y chromosome / X chromosome is bigger than Y chromosome;
non-disjunction leads to three copies of chromosome 13/trisomy 13.

35b. Outline the inheritance of hemophilia in humans. *[2 marks]*

Markscheme

sex-linked/on X chromosome;
recessive allele / Xh;
more common in males than females;
heterozygous females are carriers / only females can be carriers;

35c. Using an example, describe polygenic inheritance. *[3 marks]*

Markscheme

more than one gene contribute to/control same characteristic;
as number of genes increase so does possible number of phenotypes;
leads to continuous variation;
specific example; *(eg human skin color (due to differing amounts of melanin))*
*Award **[2 max]** for general points with no example.*

36a. State the name given to the situation where two alleles of a gene persist indefinitely in a population. *[1 mark]*

Markscheme

balanced polymorphism

36b. CF is a recessive condition that affects approximately 1 in 2500 births in Australia. Calculate the percentage of heterozygous individuals in the population. Show your calculation. *[2 marks]*

Markscheme

$2pq = (2)(0.98)(0.02) = 0.039$;

3.9%;

*Award **[1]** for working and **[1]** for correct answer.*

36c. Using CF as an example, distinguish between *allele frequency* and *gene pool*. *[2 marks]*

Markscheme

gene pool is all of the genetic information/genes/alleles present (in an interbreeding population);

allele frequency is the proportion of one allele of one/CF gene in a population;

37a. In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. *[1 mark]*

Identify the genotypes of the soybean plants with high oil content and four seeds in a pod that were used in the cross.

Markscheme

HhFf HhFf / (both) HhFf;

37b. In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. *[2 marks]*

Determine the genotypes of the gametes and offspring using a Punnett grid.

Markscheme

gametes	HF	Hf	hF	hf
HF	HHFF	HHFf	HhFF	HhFf
Hf	HHFf	HHff	HhFf	Hhff
hF	HhFF	HhFf	hhFF	hhFf
hf	HhFf	Hhff	hhFf	hhff

all gametes shown correctly on Punnett grid;
all offspring genotypes correct;

37c. In a strain of soybeans, high oil content (H) in seeds is dominant to low oil content (h) and four seeds in a pod (F) is dominant to two seeds in a pod (f). A farmer crosses two soybean plants, both with high oil content and four seeds in a pod. The offspring have a phenotypic ratio of 9 : 3 : 3 : 1. *[2 marks]*

Identify the phenotypes of each part of the phenotypic ratio.

Ratio	Phenotypes
9	
3	
3	
1	

Markscheme

	ratio	*phenotypes*
a.	*9*	high oil four seeds;
b.	*3*	high oil two seeds;
c.	*3*	low oil four seeds;
d.	*1*	low oil two seeds;

*Award **[1]** for any **two** correct phenotypes.*

37d. Deduce the reason for the person developing as a female. *[1 mark]*

Markscheme

no Y chromosome.

37e. Determine, with a reason, whether this karyotype shows that non-disjunction has occurred. *[1 mark]*

Markscheme

yes as there is only one X chromosome/chromosome missing/only 45 chromosomes

38. Define *gene pool*. *[1 mark]*

Markscheme

all of the genes/alleles in an interbreeding population

39a. State a function of each of the following parts of the human brain. *[2 marks]*

(i) Cerebellum

(ii) Hypothalamus

Markscheme

(i) (coordinates) unconscious motor functions/balance and movement

(ii) (maintains) homeostasis/thermoregulation/appetite/thirst / coordinates endocrine systems / secretes hormones/regulating factors

39b. Identify the **two** most closely related organisms. *[1 mark]*

Markscheme

(common) chimpanzee and bonobo

39c. Identify the species to which the Bonobo is most distantly related. *[1 mark]*

Markscheme

gibbon

39d. Describe **one** type of barrier that may exist between gene pools. *[3 marks]*

Markscheme

named barrier;
description of its action;
results in terms of gene pools;
e.g.:
behavioural barrier;
different populations mate at different times of year thus preventing interbreeding;
allele frequencies become different in the two gene pools/separates gene pools / sympatric speciation;

40a. Identify the highest and lowest probabilities of breeding for individuals of the same variety from different lakes. *[1 mark]*

Highest probability: .....

Lowest probability: .....

Markscheme

Highest probability: 0.58 *(Allow answers from 0.57–0.59)*
Lowest probability: 0.25 *(Allow answers from 0.24–0.26)*
Both required for the mark.

40b. Identify the breeding combination that results in the lowest probability of breeding. *[1 mark]*

Markscheme

different varieties from same lake / I

40c. Analyse the probability of breeding between individuals from the same lake. *[2 marks]*

Markscheme

individuals are more likely to breed if they are the same variety / individuals of different varieties have a low probability of breeding;
the probability of breeding between individuals of the same variety shows a large range of values / narrow range if of different variety;
the probability of breeding between any two individuals is always less than 0.6/correct numerical value;

40d. Scientists concluded that speciation is taking place in these populations. Discuss the evidence for speciation provided by the data. *[3 marks]*

Markscheme

data provides (strong) evidence for reproductive isolation between the varieties in each lake;
different sizes/feeding habits/habitat (shore versus open water) seem to contribute (strongly) to low breeding probability;
this could lead to speciation/formation of separate species in each lake;
same varieties from different lakes do not show strong reproductive isolation/ geographical isolation is a weak factor in speciation / no evidence of allopatric speciation;
sympatric speciation seems to be taking place because different varieties from the same lake have a low probability of breeding;

41a. Identify the highest and lowest probabilities of breeding for individuals of the same variety from different lakes. *[1 mark]*

Highest probability:

Lowest probability:

Markscheme

Highest probability: 0.58 (*Allow answers from 0.57–0.59*)
Lowest probability: 0.25 (*Allow answers from 0.24–0.26*)
Both required for the mark.

41b. Identify the breeding combination that results in the lowest probability of breeding. *[1 mark]*

Markscheme

different varieties from same lake / I

41c. Analyse the probability of breeding between individuals from the same lake. *[2 marks]*

Markscheme

individuals are more likely to breed if they are the same variety / individuals of different varieties have a low probability of breeding;
the probability of breeding between individuals of the same variety shows a large range of values / narrow range if of different variety;
the probability of breeding between any two individuals is always less than 0.6/correct numerical value;

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Markscheme

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same varieties from different lakes do not show strong reproductive isolation / geographical isolation is a weak factor in speciation / no evidence of allopatric speciation;
sympatric speciation seems to be taking place because different varieties from the same lake have a low probability of breeding;

42a. Outline the process of adaptive radiation. *[3 marks]*

Markscheme

varied members of a single species occupy a variety of niches / migration of a species to an area with a variety of niches;
natural selection/selection pressure will be different in various niches causing adaptation of groups to the varied niches;
results in many species from one ancestral species;
reproductive isolation enhances adaptive radiation;
adaptive radiation results in speciation;

42b. There has been a change of thinking; moving from gradualism to punctuated equilibrium demonstrates the changing nature of science. Discuss these two ideas about the pace of evolution. *[4 marks]*

Markscheme

in gradualism evolution occurs at a constant pace;
fossil records of gradual change with intermediate forms support this theory;
evolution of modern horse/another suitable example seems to support this view;
in punctuated equilibrium evolution proceeds rapidly for short periods of time intermittent with long periods of little change/stability;
gaps in the fossil record/lack of intermediate forms support the idea of punctuated equilibrium;
strata in the fossil record with appearance of many new species following a mass extinction supports the idea of punctuated equilibrium;

43a. Compare sympatric speciation and allopatric speciation. *[2 marks]*

Markscheme

both involve reproductive isolation / separation of gene pools;

sympatric is speciation due to isolation of populations living in the same geographic area whereas allopatric is speciation due to geographic isolation;

43b. Discuss the concept of punctuated equilibrium. *[3 marks]*

Markscheme

long periods of stability;

environmental change / sudden change;

leads to rapid speciation/evolution;

helps to explain absence of intermediate forms;

competing theory is gradualism;

44a. State the genotype of the heterozygous parent using the correct notation. *[1 mark]*

Markscheme

$\frac{T\ \ E}{t\ \ e}$

44b. Identify which individuals are recombinants in this cross. *[1 mark]*

Markscheme

unenclosed seeds, starchy and tunica present, sugary /

$\frac{T\ \ e}{t\ \ e}$ and $\frac{t\ \ E}{t\ \ e}$ *(both needed)*

44c. Explain what has occurred to cause these results. *[2 marks]*

Markscheme

crossing over;
between non-sister chromatids (in prophase I);
results in exchange of alleles / change in linkage groups;
so some gametes are T e or t E; (*linkage notation not expected*)
test cross expect ratio of two phenotypes / correct Punnett Square showing test cross;
but instead get four phenotypes with smaller percentage of recombinants;
Above points can be shown in diagrams.

44d. Maize belongs to the group of plants known as angiospermophyta. Distinguish between angiospermophytes and bryophytes. *[2 marks]*

Markscheme

angiospermophytes	bryophytes
flowering	non-flowering;
(true) roots	rhizoids/no true roots;
(true) leaves	scales / thallus/no true leaves;
seeds produced	spores produced (in capsule);
waxy cuticle	no cuticle;
vascular (tissue)	non-vascular / no vascular tissue

45a. Explain, using a **named** example, how polygenic inheritance gives rise to continuous variation. *[2 marks]*

Markscheme

human skin colour can vary from pale to very dark / amount of melanin varies;
skin colour/melanin controlled by (alleles from) at least three/several genes;
no alleles are dominant / alleles are co-dominant / incomplete dominance;
many different possible combinations of alleles;
skin colour controlled by cumulative effect/combination of genes/alleles;
Award the above marking points for any other valid example.

45b. Describe the inheritance of colour blindness in humans. *[3 marks]*

Markscheme

sex linked condition;
carried on an X chromosome / absent from Y chromosome;
if present in male causes colour blindness;
(allele is) recessive so heterozygous females are not colour blind;
homozygous females are colour blind;
Do not allow carried on sex chromosome.

46. Discuss evolution by gradualism and punctuated equilibrium. *[6 marks]*

Markscheme

a. both describe the pace/speed/rate of evolution;

b. gradualism suggests that evolution occurs over a long time;

c. gradualism changes are slow/steady over time;

d. gradualism would occur when there is little change in the environment;

e. punctuated equilibrium implies long periods with no change;

f. punctuated equilibrium implies short periods with great change;

g. punctuated equilibrium occurs when there are great changes in the environment;

h. example; *(eg: in times of volcanic activity/meteorite impact/great climate change / OWTTE)*

i. generally accepted that both ideas take place in evolution

47a. Outline the processes that occur during the first division of meiosis. *[6 marks]*

Markscheme

Remember, up to TWO "quality of construction" marks per essay.

a. (consists of) prophase, metaphase, anaphase and telophase;
b. chromosome number halved/reduced/(diploid) to haploid;
c. homologous chromosomes pair up/form a bivalent/synapsis in prophase;
d. crossing over between non-sister chromatids/chromatids of different homologues;
e. nuclear envelope breaks down (at end of prophase/start of metaphase);
f. tetrads/bivalents/homologous pairs move to/align on equator/cell centre/on metaphase plate in metaphase; (accept homologous chromosomes without pairs if pairing has already been described)
g. attachment of spindle fibres/microtubules to centromeres/kinetochores;
h. (homologous) chromosomes separate/pulled to opposite poles in anaphase;
i. nuclear envelopes reform/do not reform (because of meiosis II) in telophase;
Accept the above points in a series of annotated diagrams. Reject answers with single chromatids forming pairs in metaphase or separating or moving to opposite poles in anaphase.

47b. Prior to cell division, chromosomes replicate. Explain the process of DNA replication in prokaryotes. *[8 marks]*

Markscheme

Remember, up to TWO "quality of construction" marks per essay.

a. DNA replication is semi-conservative;
b. each (molecule formed) has one new strand and one from parent molecule;
c. helicase uncoils DNA;
d. helicase separates the two strands by breaking hydrogen bonds between bases; (reject unzips as an alternative to uncoils but accept as alternative to separates if breakage of hydrogen bonds is included)
e. RNA primase adds primer / primase adds (short) length of RNA;
f. DNA polymerase III binds to/starts at (RNA) primer;
g. DNA polymerase (III) adds nucleotides/bases in a 5' → 3' direction;
h. bases according to complementary base pairing / A–T and C–G;
i. (leading strand) built up continuously (towards the replication fork);
j. (lagging strand) built up in pieces/short lengths/Okazaki fragments;
k. DNA polymerase I removes RNA/primers and replaces them with DNA;
l. ligase seals gaps between nucleotides/fragments/makes sugar-phosphate bonds;
m. nucleoside triphosphates provide the energy to add nucleotides;
Accept the above points in annotated diagrams.

47c. Outline outcomes of the human genome project. *[4 marks]*

Markscheme

Remember, up to TWO "quality of construction" marks per essay.

a. complete human DNA/chromosomes sequenced;
b. identification of all human genes / find position/map (all) human genes;
c. find/discover protein structures/functions;
d. find evidence for evolutionary relationships/human origins/ancestors;
e. find mutations/base substitutions/single nucleotide polymorphisms;
f. find genes causing/increasing chance of/develop test for/screen for diseases;
g. develop new drugs (based on base sequences) / new gene therapies;
h. tailor medication to individual genetic variation / pharmacogenomics;
i. promote international co-operation/global endeavours;

48a. Describe the causes of Down syndrome. *[5 marks]*

Markscheme

Down syndrome is caused by non-disjunction;
occurs during meiosis;
chromosome pairs fail to separate in meiosis I / chromatids in meiosis II / anaphase II;
some gametes have an extra chromosome;
can lead to zygotes/individuals with an extra chromosome / individual has 47 chromosomes;
in Down syndrome this would be trisomy 21/extra chromosome 21;
increased probability with increased age of mother/ages of parents;

48b. Describe how human skin colour is determined genetically. *[5 marks]*

Markscheme

skin colour is an example of polygenic inheritance;
many/more than two genes contribute to a person's skin colour;
due to the amount of melanin in the skin;
combination of alleles determines the phenotype;
allows for range of skin colours / continuous variation of skin colour;
phenotypes do not follow simple Mendelian ratios of dominance and recessiveness;
the environment also affects gene expression of skin colour / sunlight/UV light stimulate melanin production;
the more recessive alleles there are, the lighter the skin colour; (*vice versa*)

48c. Explain the causes of sickle-cell anemia. *[8 marks]*

Markscheme

caused by gene mutation;
(sickle-cell anemia) due to a base substitution (mutation);
changes the code on the DNA;
which leads to a change in transcription / change in mRNA;

DNA changes from CTC to CAC/GAG to GTG / mRNA changes from GAG to GUG; (*accept DNA changes from CTT to CAT/GAA to GTA / mRNA changes from GAA to GUA*)

which (in turn) leads to a change in translation / change in polypeptide chain/ protein;
(the tRNA) adds the wrong amino acid to the polypeptide chain;
glutamic acid replaced by valine;
produces abnormal hemoglobin;
causing abnormal red blood cell/erythrocyte shape / sickle shape;
which lowers the ability to transport oxygen;
sickle-cell allele is codominant;
homozygote/Hb^S Hb^S have sickle cell anemia/is lethal / heterozygote/Hb^S Hb^A has the sickle trait/is carrier (and is more resistant to malaria);

49a. Draw a labelled diagram of a mature sperm. *[5 marks]*

Markscheme

*Award **[1]** for each of the following clearly drawn and correctly labelled.*
head and midpiece/mid-section/body;
tail/flagellum; (*at least four times length of the head and containing fibres*)
acrosome; (*shown as distinct structure near front of head*)
nucleus; (*occupying more than half the width or length of head*)
mitochondria; (*as repetitive structures inside membrane of mid piece*)
centriole; (*between head and midpiece*)
(plasma) membrane; (*shown as single line covering whole cell*)
microtubules; (*in 9 plus 2 array*)

49b. Outline the formation of chiasmata during crossing over. *[5 marks]*

Markscheme

crossing over/chiasmata formed during prophase I of meiosis;
pairing of homologous chromosomes/synapsis;
chromatids break (at same point); (*do not accept chromatids overlap*)
non-sister chromatids join up/swap/exchange alleles/parts;
X-shaped structure formed / chiasmata are X-shaped structures;
chiasma formed at position where crossing over occurred;
chiasmata become visible when homologous chromosomes unpair;
chiasma holds homologous chromosomes together (until anaphase);
Accept the above points in an appropriately annotated diagram.

49c. Explain how an error in meiosis can lead to Down syndrome. *[8 marks]*

Markscheme

non-disjunction;
chromosomes/chromatids do not separate / go to same pole;
non-separation of (homologous) chromosomes during anaphase I;
due to incorrect spindle attachment;
non-separation of chromatids during anaphase II;
due to centromeres not dividing;
occurs during gamete/sperm/egg formation;
less common in sperm than egg formation / function of parents' age;
Down syndrome due to extra chromosome 21;
sperm/egg/gamete receives two chromosomes of same type;
zygote/offspring with three chromosomes of same type / trisomy / total 47 chromosomes;
Accept the above points in an appropriately annotated diagram.