Past-paper walk-through

Non-Mendelian Genetics ▪ 5.4

eXercise

Questions in this set

- 2025 Q6
- 2023 Q2
- 2021 Q2
- 2019 Q3
- 2019 Q6
- 2018 Q7
- 2016 Q7

Reading the mark scheme

- **B1** a mark that stands on its own – the point is either made or not.
- **M1** a *method* mark: the right approach, even if the number is wrong.
- **A1** an *answer* mark, and it usually needs its M mark first.
- **C1** a working mark on the way to the answer.
- // separates answers the examiner accepts equally.
- **ecf** = error carried forward: a later part is marked on your own earlier answer.
- **owtte** = "or words to that effect" – the wording need not match.

Question 6

2025 ▪ Q6

The *ald* gene of fruit flies encodes the ALD protein, which is associated with both the centromeres of chromosomes and protein filaments produced during meiosis. In the absence of functional ALD proteins, gamete-producing cells enter anaphase I before homologous chromosomes are correctly aligned. As a result, the gametes produced do not contain the correct numbers of chromosomes.

Scientists generated four mutations in the *ald* gene: *ald1*, *ald3*, *ald23*, and *del*, which was a deletion of the gene. To study the role of the ALD protein in meiosis, scientists used gameteforming metaphase cells from groups of flies with different *ald* genotypes. Some of the flies were homozygous for the wild-type allele of *ald*: *WT/WT*. Other flies were heterozygous for different *ald* alleles: *WT/del*; *ald1/del*; *ald3/ald23*; *ald23/del*. The scientists measured the

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percent of metaphase cells that contained ALD-associated filaments (Figure 1A) and the amount of ALD protein produced by each of the cell types (Figure 1B).
Figure 1. (A) The average percent of gamete-forming metaphase cells that contained filaments associated with ALD and (B) the amount of ALD protein produced by each cell type. A thicker band indicates a greater amount of ALD protein.
[Panel A: Bar graph; y-axis "Percent of Metaphase Cells with ALD-Associated Filaments" from 0 to 100 in increments of 20. x-axis "*ald* Genotype" with five bars labeled "WT/WT", "WT/del", "ald1/del", "ald3/ald23", "ald23/del". Approximate values with error bars: WT/WT ≈ 63 (error bar roughly 45-80), WT/del ≈ 77 (error bar roughly 65-88), ald1/del ≈ 13 (error bar roughly 5-20), ald3/ald23 ≈ 6 (small error bar), ald23/del ≈ 1 (negligible, near 0).
Panel B: Gel-blot style diagram labeled "ALD Protein" showing bands of varying thickness for the same five genotypes in the same order (WT/WT, WT/del, ald1/del,

Question 6

ald3/ald23, ald23/del): WT/WT shows a thick solid band; WT/del shows a medium-thick band; ald1/del shows a medium-thick band similar to WT/del; ald3/ald23 shows a thin band; no band is shown for ald23/del (or an extremely faint/absent line).]

(a) Based on Figure 1A, identify the fly genotype in which the average percent of metaphase cells with ALD-associated filaments is close to 12%.

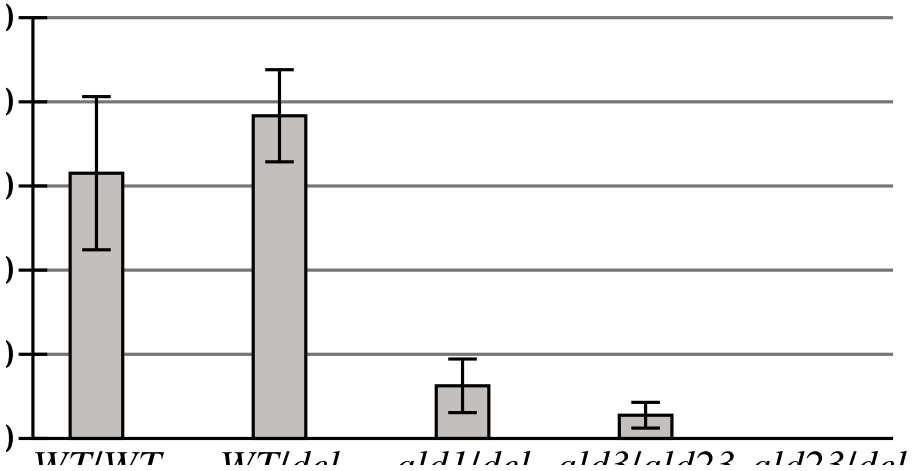
(b) Based on Figure 1B, describe the difference in ALD protein production between gamete-forming metaphase cells of flies with the genotype *ald3/ald23* and flies with the genotype *ald23/del*.

(c) Scientists hypothesize that gamete-forming metaphase cells can produce a normal amount of ALD-associated filaments even when they produce about half as much ALD protein as the wild-type cells produce. Use the data in Figures 1A and 1B to support the scientists' hypothesis.

Question 6

(d) For gamete-forming metaphase cells of the *WT/del* and *ald1/del* flies, explain why the phenotypes observed in Figure 1A differ even though the amount of ALD protein produced (Figure 1B) does not.

Question 6



Solution 6

(a)

Mark scheme

- The genotype ald1/del has an average of about 12% of metaphase cells with ALD-associated filaments, based on Figure 1A.

Solution 6

(b)

Mark scheme

- More ALD protein is produced by ald3/ald23 cells than by ald23/del cells (Figure 1B shows a thin but visible band for ald3/ald23, while ald23/del shows little to no detectable band) — equivalently, ald23/del cells produce little to no ALD protein compared with ald3/ald23 cells.

Solution 6

(c)

Mark scheme

- The WT/del cells produce about half as much ALD protein as WT/WT cells (Figure 1B, a thinner band), yet show no substantial difference from WT/WT in the percent of metaphase cells with ALD-associated filaments (Figure 1A, roughly 76% vs 63%, with overlapping error bars) — this comparison supports the hypothesis that gamete-forming metaphase cells can produce a normal amount of ALD-associated filaments with about half the normal ALD protein.

Solution 6

(d)

Mark scheme

- The phenotypes differ because it is protein function, not quantity, that differs: WT/del flies produce ALD protein from their functional WT allele, which is enough to generate a wild-type phenotype (~76% of cells with filaments), whereas ald1/del flies produce a similar total amount of ALD protein (Figure 1B) but it comes from the nonfunctional/impaired ald1 allele, so it cannot properly support filament formation, giving a much lower percentage of cells with ALD-associated filaments (~12%, Figure 1A).

Question 2

2023 ▪ Q2

Elevated levels of CO_2 increase the rate of photosynthesis and growth in plants. Scientists studying the mechanisms involved in these increases examined a variety of species and found that when plants are exposed to elevated levels of CO_2, there is an increase in the number of chloroplasts per cell. To investigate whether the elevated levels of CO_2 have a similar effect on the number of mitochondria in plant cells, the scientists then selected six of these species to quantify the number of mitochondria per cell when the plants were exposed to both normal and elevated levels of CO_2 (Table 1).
TABLE 1. AVERAGE NUMBER OF MITOCHONDRIA IN PLANTS EXPOSED TO NORMAL AND ELEVATED LEVELS OF CO_2
| Species | Mitochondria at Normal CO_2 (per 100 μm^2 of cell area) $\pm 2SE_{\bar{x}}$ | Mitochondria at Elevated CO_2 (per 100 μm^2 of cell area) $\pm 2SE_{\bar{x}}$ | |—|—|—| | 1 |

Question 2

1.0 ± 0.10 | 1.6 ± 0.10 | | 2 | 0.4 ± 0.05 | 0.9 ± 0.08 | | 3 | 0.5 ± 0.07 | 0.9 ± 0.10 | | 4 | 0.3 ± 0.03 | 0.6 ± 0.06 | | 5 | 0.7 ± 0.06 | 1.5 ± 0.22 | | 6 | 1.3 ± 0.15 | 2.4 ± 0.22 |

(a) Describe the role of the inner mitochondrial membrane in cellular respiration.

Question 2

2023 ▪ Q2

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3	0.5 ± 0.07	0.9 ± 0.10
4	0.3 ± 0.03	0.6 ± 0.06
5	0.7 ± 0.06	1.5 ± 0.22
6	1.3 ± 0.15	2.4 ± 0.22

Question 2

(b) Using the template in the space provided for your response, **construct** an appropriately labeled graph that represents the data in Table 1. **Determine** which species show(s) a difference in the number of mitochondria between normal and elevated levels of CO_2.

Question 2

2023 ▪ Q2

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Question 2

(c) Based on the data in Table 1, **describe** the relationship between the level of CO_2 and the average number of mitochondria per unit area of a cell.

Question 2

2023 ▪ Q2

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Question 2

(d) The leaves of a particular plant species are typically green, but scientists notice a plant in which the leaves have white stripes. They determine that the stripes result from a mutation in mitochondrial DNA that interferes with the development of chloroplasts. The scientists crossed plants using pollen from the plant with white-striped leaves and ovules from a plant with green leaves. **Predict** the phenotype(s) of the leaves of offspring produced from this cross. Provide reasoning to **justify** your prediction. **Explain** why plants with the same genotype are able to differ in the structure and/or number of certain organelles in response to changes in atmospheric levels of CO_2.

Solution 2

(a)

Mark scheme

- The inner mitochondrial membrane houses the electron transport chain components and ATP synthase, i.e., it is the site of oxidative phosphorylation. By separating the intermembrane space from the matrix, it allows a proton ($H+$) gradient to be established across it as electrons move through the ETC; this gradient then drives ATP synthesis via chemiosmosis through ATP synthase.

Solution 2

(b) Mark scheme

- Construct a bar graph (or modified/grouped bar graph) with the six plant species (1–6) on the x-axis and average number of mitochondria (per 100 μm^2 of cell area) on the y-axis, with appropriate axis labels/units and a legend distinguishing normal-CO2 bars from elevated-CO2 bars. Plot each species' two data points (normal and elevated CO2) at the values given in Table 1, each with error bars representing $\pm 2SE(\bar{x})$ as listed in the table (e.g., species 1: 1.0 ± 0.10 normal, 1.6 ± 0.10 elevated; species 6: 1.3 ± 0.15 normal, 2.4 ± 0.22 elevated). Points earned for: appropriate labeling; using a bar/modified bar graph format; correctly plotting the data points and error bars. Determine which species show a difference in mitochondria number between normal and elevated CO2: all six species (1

Solution 2

through 6) show a difference — every species has a higher (non-overlapping, given the error bars) mitochondria count under elevated CO2 than under normal CO2.

Solution 2

(c)

Mark scheme

- There is a positive relationship between CO2 level and the average number of mitochondria per unit cell area: the number of mitochondria is greater under elevated CO2 conditions than under normal CO2 conditions (across the species examined).

Solution 2

(d) Mark scheme

- Prediction: the offspring from this cross (pollen from the white-striped-leaf plant $\times$ ovules from the green-leaf plant) will all have green leaves (will not have white stripes). Justification: mitochondria (and mitochondrial DNA) are maternally inherited — they are transferred to offspring through the ovule/egg cytoplasm, not through the pollen — so all offspring inherit the mitochondrial genotype of the green (ovule-producing) parent, whose mitochondria are functional for chloroplast development. Separately, explain why plants with the same (nuclear) genotype can still differ in the structure and/or number of certain organelles in response to CO2: because changes in CO2 levels (the environment) affect the expression of certain genes — i.e., environmentally-induced differences in gene expression (not

Solution 2

differences in genotype) drive differences in organelle structure/number, such as the increase in mitochondria number seen at elevated CO2.

Question 2

2021 ▪ Q2

Geneticists investigated the mode of inheritance of a rare disorder that alters glucose metabolism and first shows symptoms in adulthood. The geneticists studied a family in which some individuals of generations II and III are known to have the disorder. Based on the pedigree (Figure 1), the geneticists concluded that the disorder arose in individual II−2 and was caused by a mutation in mitochondrial DNA.

[Figure 1: Pedigree diagram across four generations (I, II, III, IV). Generation I: individual 1 (female without disorder) married to individual 2 (male without disorder). Generation II: their children include individual 1 (male without disorder) married to individual 2 (female with disorder), individual 3 (male without disorder) married to individual 4 (female without disorder), and individual 5 (female without disorder) married to individual 6 (male without disorder). Generation III: children of II-1×II-2 are individual 1 (male without disorder) married to individual 2 (female with disorder),

Question 2

individual 3 (female without disorder) married to individual 4 (male with disorder), individual 5 (male without disorder) married to individual 6 (female with disorder), and individual 7 (female without disorder) married to individual 8 (male with disorder); children of II-3×II-4 are individual 9 (female without disorder) and individual 10 (male without disorder) married to individual 11 (female without disorder); children of II-5×II-6 are individual 12 (male without disorder), individual 13 (female without disorder), individual 14 (female without disorder). Generation IV: child of III-1×III-2 is individual 1 (female, unknown phenotype); children of III-3×III-4 are individuals 2 and 3 (both female, unknown phenotype); children of III-5×III-6 are individuals 4 and 5 (both female, unknown phenotype); children of III-7×III-8 is individual 6 (male, unknown phenotype); children of III-10×III-11 are individual 7 (female, unknown phenotype) and individual 8 (male, unknown phenotype). Legend: open circle = female without disorder, filled circle = female with disorder, circle with "?" = female

Question 2

with unknown phenotype; open square = male without disorder, filled square = male with disorder, square with "?" = male with unknown phenotype. Caption: "Figure 1. Pedigree of a family showing individuals with the glucose metabolism disorder. A question mark indicates that the phenotype is unknown."]

TABLE 1. AVERAGE BLOOD GLUCOSE LEVELS OF INDIVIDUALS IN GENERATION IV

| Individual | Average Blood Glucose Level (mg/dL $\pm$ 2SE$_{\bar{x}}$) | |—|—| | IV−1 | 170 $\pm$ 15 | | IV−2 | 190 $\pm$ 10 | | IV−3 | 145 $\pm$ 5 | | IV−4 | 165 $\pm$ 15 | | IV−5 | 110 $\pm$ 15 | | IV−6 | 125 $\pm$ 5 | | IV−7 | 105 $\pm$ 15 | | IV−8 | 120 $\pm$ 10 |

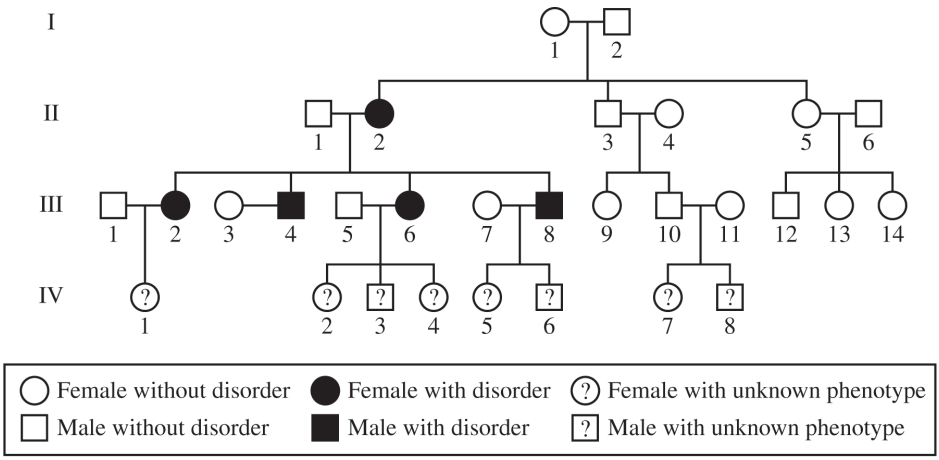
TABLE 2. PHENOTYPIC CLASSIFICATIONS BASED ON BLOOD GLUCOSE LEVELS

| Phenotype | Blood Glucose Level (mg/dL) | |—|—| | Normal | $<$ 140 mg/dL | | At risk | 140 $-$ 199 mg/dL | | Affected | $\geq$ 200 mg/dL |

Question 2

(a) The disorder alters glucose metabolism. **Describe** the atoms AND types of bonds in a glucose molecule.
(b) Using the template in the space provided for your response, **construct** an appropriately labeled graph based on the data in Table 1. **Determine** one individual who is both at risk of developing the disorder and has a significantly different blood glucose level from that of individual IV−1.
(c) Based on the pedigree, **identify** all individuals in generation IV who can pass on the mutation to their children.
(d) Based on the fact that individual II−2 is affected, a student claims that the disorder is inherited in an X-linked recessive pattern. Based on the student's claim, **predict** which individuals of generation III will be affected by the disorder. Based on the pedigree, **justify** why the data do NOT support the student's claim.

Question 2


I
II
III
IV
Female without disorder
Female with disorder
Female with unknown phenotype
Male without disorder
Male with disorder
Male with unknown phenotype

Solution 2

(a)

Mark scheme

- The atoms in a glucose molecule are carbon, hydrogen, and oxygen (C, H, and O), held together by covalent bonds.

Solution 2

(b) Mark scheme

- Construct graph (3 pts, point distribution across: axis labels; plotting in a bar graph or modified bar graph; error bars): Plot a bar graph with Individual (IV-1 through IV-8) labeled on the x-axis and Average Blood Glucose Level (mg/dL) labeled on the y-axis, one bar per individual at the height given in Table 1 (IV-1=170, IV-2=190, IV-3=145, IV-4=165, IV-5=110, IV-6=125, IV-7=105, IV-8=120), each with error bars matching its $\pm$2SE value from Table 1 (15,10,5,15,15,5,15,10 respectively). Determine (1 pt): IV-3 is both at risk (blood glucose 145 mg/dL falls in the 140-199 'at risk' range per Table 2) and has an average blood glucose level significantly different from individual IV-1 (170$\pm$15 vs 145$\pm$5 — the error bars/2SE ranges do not overlap).

Solution 2

(c)

Mark scheme

- The individuals in generation IV who can pass on the mutation to their children are IV-1, IV-2, and IV-4 (their mother, III-6, is an affected female; because the disorder is due to a mitochondrial DNA mutation transmitted maternally, all of III-6's offspring inherit and can pass on the mutation, regardless of the offspring's own phenotype/sex).

Solution 2

(d)

Mark scheme

- Predict affected individuals under the X-linked recessive claim (1 pt): III-4 and III-8 would be predicted affected in generation III. Justify why the data do NOT support the claim (1 pt): Accept either — the data do not support the claim because females III-2 and III-6 have the disorder, and under X-linked recessive inheritance they could only be affected if their father II-1 had the disorder, which he does not; or the data instead support mitochondrial inheritance, because all of the offspring of individual II-2 (affected female), not only the sons, have the disorder.

Question 3

2019 ▪ Q3

The pyruvate dehydrogenase complex (PDC) catalyzes the conversion of pyruvate to acetyl-CoA, a substrate for the Krebs (citric acid) cycle. The rate of pyruvate conversion is greatly reduced in individuals with PDC deficiency, a rare disorder.

(a) Identify the cellular location where PDC is most active.

(b) Make a claim about how PDC deficiency affects the amount of NADH produced by glycolysis AND the amount of NADH produced by the Krebs (citric acid) cycle in a cell. **Provide reasoning** to support your claims based on the position of the PDC-catalyzed reaction in the sequence of the cellular respiration pathway.

(c) PDC deficiency is caused by mutations in the *PDHA1* gene, which is located on the X chromosome. A male with PDC deficiency and a homozygous female with no family history of PDC deficiency have a male offspring. **Calculate** the probability that the male offspring will have PDC deficiency.

Solution 3

(a)

Mark scheme

- PDC (pyruvate dehydrogenase complex) is most active in the mitochondria, specifically the mitochondrial matrix, where pyruvate is converted to acetyl-CoA before entering the Krebs cycle.

Solution 3

(b) Mark scheme

- Glycolysis: Claim — no change in the amount of NADH produced by glycolysis. Reasoning — glycolysis occurs in the cytoplasm, before (upstream of) the PDC-catalyzed conversion of pyruvate to acetyl-CoA, so glycolysis continues normally and does not require PDC; its NADH output is unaffected. [1 point] Krebs (citric acid) cycle: Claim — decrease in the amount of NADH produced by the Krebs cycle. Reasoning — the Krebs cycle occurs after (downstream of) the PDC-catalyzed step in the pathway, using acetyl-CoA as its substrate; with PDC deficiency there is little/no acetyl-CoA available, so the Krebs cycle (and the NADH it generates) is greatly reduced/slowed down. [1 point]

Solution 3

(c)

Mark scheme

- The probability that the male offspring will have PDC deficiency is 0 (0%). The PDHA1 gene is X-linked. A male offspring receives his single X chromosome from his mother and his Y chromosome from his father. The mother is homozygous with no family history of the disorder, so both of her X alleles are normal (non-mutant); she can only pass a normal X to her son. The father's mutant X (which caused his own PDC deficiency) is never passed to a son (sons get his Y). Therefore the offspring cannot/will not have PDC deficiency — probability = 0.

Question 6

2019 ▪ Q6

TABLE 1 (data table referenced by the question, reproduced below as Table 1 with the growth results for the three haploid strains):

	MEDIUM	STRAINS — Wild Type	STRAINS — Mutant 1	STRAINS — Mutant 2
Treatment I	All amino acids present	+	+	+
Treatment II	No amino acids present	+	−	−
Treatment III	All amino acids present EXCEPT methionine	+	−	+
Treatment IV	All amino acids present EXCEPT leucine	+	+	−

Question 6

Table 1. The data show the growth of haploid *Saccharomyces cerevisiae* yeast strains on media that differ in amino acid content. A plus sign (+) indicates that the yeast strains grow, and a minus sign (−) indicates that the strains do not grow.
The yeast *Saccharomyces cerevisiae* is a single-celled organism. Amino acid synthesis in yeast cells occurs through metabolic pathways, and enzymes in the synthesis pathways are encoded by different genes. The synthesis of a particular amino acid can be prevented by mutation of a gene encoding an enzyme in the required pathway.

Question 6

A researcher conducted an experiment to determine the ability of yeast to grow on media that differ in amino acid content. Yeast can grow as both haploid and diploid cells. The researcher tested two different haploid yeast strains (Mutant 1 and Mutant 2), each of which has a single recessive mutation, and a haploid wild-type strain. The resulting data are shown in Table 1.

(a) Identify the role of treatment I in the experiment.

Question 6

		STRAINS			
	MEDIUM	Wild Type (haploid)	Mutant 1 (haploid)	Mutant 2 (haploid)	Diploid Cells Produced by Mating Mutant 1 and Mutant 2
Treatment I	All amino acids present	+	+	+	
Treatment II	No amino acids present	+	–	–	
Treatment III	All amino acids present EXCEPT methionine	+	–	+	
Treatment IV	All amino acids present EXCEPT leucine	+	+	–	

Question 6

		STRAINS		
	MEDIUM	Wild Type	Mutant 1	Mutant 2
Treatment I	All amino acids present	+	+	+
Treatment II	No amino acids present	+	−	−
Treatment III	All amino acids present EXCEPT methionine	+	−	+
Treatment IV	All amino acids present EXCEPT leucine	+	+	−