1: genes, environment, clinical heterogeneity

Question	Marks	Question type
i		Information or resources
1.1	1	Multiple Choice
1.2	1	Multiple Choice
1.3	1	Multiple Choice
1.4	1	Multiple Choice
1.5	3	Essay
1.6	3	Essay
1.7	2	Essay

2: pharmagogenetics, individualized medicine

Question	Marks	Question type
2.1	1	Multiple Choice
2.2	1	Multiple Choice
2.3	1	Multiple Choice
2.4	1	Multiple Choice
2.5	1	Essay
2.6	2	Essay
2.7	2	Essay
2.8	2	Essay
2.9	1	Essay

3: functional genetics

Question	Marks	Question type
3.1	1	Multiple Choice
3.2	1	Multiple Choice
3.3	1	Multiple Response

	3.4	1	Multiple Choice
	3.5	3	Essay
	3.6	2	Essay
	3.7	3	Essay

1.5 Explain locus heterogeneity using the case of two patients with retinitis pigmentosa (RP), one with an RPGR mutation and the other with a RHO mutation, yet similar clinical presentations. Discuss diagnostic and therapeutic implications. (3p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 3

1.6 A pair of identical twins are diagnosed with Type 1 diabetes. One twin experiences severe complications, while the other maintains relatively stable blood sugar levels with minimal intervention. What factors could explain this clinical heterogeneity? (3p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 3

1.7 Using PKU as a model, discuss how understanding G×E interactions leads to effective prevention strategies. (2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

2.5 Define sensitivity and specificity of a screening test. (1p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 1

2.6 Phase I in drug metabolism relies largely in the P450 Cytochrome family. Explain how and why these enzymes are so important for drug metabolism (1p), their main organ of action (0,5p), and discuss the factors that can alter their activity (0,5p). (2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

2.7 Phase II metabolism consists mainly of conjugation reactions. Explain how these reactions modify drugs or metabolites (0,5p), describe how they influence drug toxicity and excretion (1p), and give one example of a Phase II pathway that shows genetic variability (0,5p). (2 p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

2.8 In recent years, several in vivo CRISPR–Cas treatments have shown promising clinical results, including therapies for sickle cell disease, and CPS1 deficiency. Using your own scientific reasoning, discuss what could potentially go wrong during such procedures and explain why caution is necessary when deploying permanent gene-editing therapies in humans. (2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

2.9 Describe the terms pharmacokinetics and pharmacodynamics (1p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 1

3.5 Briefly describe how the Cre-loxP-system works, and explain how it can be used to study gene function in a mouse model. (3p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 3

3.6 Name two unicellular model organisms that are used for genetic research, and briefly describe what type of processes can be investigated in these models. (2p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 2

3.7 Name three different categories (types) of cellular disease models, and briefly describe one specific cell model in each category. (3p)

Write your answer in the box below. Changes are saved automatically.

Format

Words: 0

Maximum marks: 3