



B6H Inheritance, Variation and Evolution: Higher Tier Practice

AQA 8461 · Calculator allowed · about 95 minutes

Total Marks

Name: _____ Date: ____ / ____ / ____

Answer ALL questions. Show all your working.

- 1** State the meaning of the terms 'homozygous' and 'heterozygous' as applied to an organism's genotype for a particular gene.

(Total for Question 1 is 2 marks)

- 2** Polydactyly (having extra fingers or toes) is caused by a dominant allele, D. The recessive allele, d, gives the usual number of digits. A man with genotype Dd (polydactyly) and a woman with genotype dd (usual number of digits) plan to have children.

Draw a Punnett square for this cross and calculate the percentage of their children expected to have polydactyly.

(Total for Question 2 is 3 marks)

3 A family pedigree is described below for an inherited condition.

Generation 1: a father who does not have the condition and a mother who does not have the condition.

Generation 2: three of their children. Two children do not have the condition. One child, a daughter, has the condition.

Use this information to answer the following.

(a) State, with a reason, whether the allele for the condition is dominant or recessive.

(b) State the genotype of the father and the mother, using F for the dominant allele and f for the recessive allele.

(Total for Question 3 is 3 marks)

4 Two parents who are both carriers of cystic fibrosis have genotype Ff, where f is the recessive allele that causes cystic fibrosis.

Calculate the percentage of their children expected to be: (a) unaffected and not a carrier (genotype FF); (b) an unaffected carrier (genotype Ff); (c) affected by cystic fibrosis (genotype ff).

(Total for Question 4 is 4 marks)

5 Explain why genetic variation within a species is important for the long-term survival of that species when the environment changes.

(Total for Question 5 is 3 marks)

6 Extended response (6 marks).

A single population of a species of fish once lived throughout a large lake. A geological event divided the lake into two separate lakes, with no way for fish to move between them, around 50,000 years ago.

Explain how this separation could lead to the formation of two different species of fish over time.

(Total for Question 6 is 6 marks)

7 An evolutionary tree is a diagram used by scientists to show how species are related through evolution. It is described in words below.

Species A and species B share a common ancestor that lived more recently than the common ancestor shared by species A, B and species C. Species D branched off earlier still, from an ancestor common to all four species.

Use this description to answer:

- (a) State which two species, A/B/C/D, are most closely related to each other.
- (b) State which species is the most distantly related to species A.

(Total for Question 7 is 2 marks)

- 8** Modern classification uses a three-domain system, first proposed by Carl Woese, which groups all living organisms into Archaea, Bacteria and Eukarya, above the traditional kingdom level.

State the type of evidence that allowed scientists to develop the three-domain system, and explain why this evidence gave more detailed information than looking at an organism's physical (visible) features alone.

(Total for Question 8 is 3 marks)

- 9** A hospital tests the bacteria on a ward for resistance to a particular antibiotic before and after a period when the antibiotic is used heavily.

Before: 40 out of 500 bacteria sampled were resistant.

After: 210 out of 500 bacteria sampled were resistant.

- (a) Calculate the percentage of resistant bacteria before and after.
- (b) Calculate the percentage point increase in resistant bacteria.
- (c) Explain, in terms of natural selection, why this increase happened.

(Total for Question 9 is 5 marks)

- 10** Human insulin can be manufactured using genetic engineering, by inserting the human insulin gene into bacteria.

Describe the main stages of this process, from isolating the human insulin gene to bacteria producing insulin.

(Total for Question 10 is 4 marks)

- 11** Selective breeding can quickly produce animals or plants with desirable characteristics, but it also has disadvantages.

Evaluate the impact of repeated selective breeding on the genetic variation of a population, and explain one risk this creates.

(Total for Question 11 is 3 marks)

- 12** A mutation changes one base in the DNA sequence of a gene that codes for an enzyme.

Explain how this single base change could result in the enzyme no longer working, linking your answer to protein structure.

(Total for Question 12 is 3 marks)

- 13** Explain how meiosis produces gametes that are genetically different from each other and from the parent cell.

(Total for Question 13 is 3 marks)

- 14** In a population of moths, a survey recorded the percentage of moths with a dark-coloured allele over three generations, as air pollution in the area decreased.

Generation 1: 92 % dark-coloured

Generation 2: 74 % dark-coloured

Generation 3: 45 % dark-coloured

- (a) Calculate the total percentage point decrease in dark-coloured moths from Generation 1 to Generation 3.
- (b) Suggest an explanation, in terms of natural selection, for this trend as pollution decreased.

(Total for Question 14 is 4 marks)

Mark scheme · B6H Inheritance, Variation and Evolution: Higher Tier Practice

Question 1

- **B1** homozygous: the two alleles for a gene are the same (both dominant or both recessive), oe
- **B1** heterozygous: the two alleles for a gene are different (one dominant and one recessive), oe
- **Answer: Homozygous: both alleles for a gene are the same. Heterozygous: the two alleles are different.**

Question 2

- **M1** correctly combines gametes D, d from the father with d, d from the mother to give genotypes Dd, Dd, dd, dd
- **M1** identifies 2 out of 4 offspring have genotype Dd (polydactyly)
- **A1** 50 % cao
- **Answer: 50 % of their children are expected to have polydactyly.**

Question 3

- **B1** the allele is recessive
- **B1** because two unaffected parents have produced an affected child, so both parents must carry a hidden copy of the allele, which can only happen if it is recessive, oe
- **B1** both parents have genotype Ff (heterozygous / carriers)
- **Answer: The allele is recessive, because two unaffected (carrier) parents produced an affected child. Both parents have genotype Ff.**

Question 4

- **M1** correctly combines gametes F, f from each parent to give genotypes FF, Ff, Ff, ff
- **B1** (a) 25 % (1 in 4) FF cao
- **B1** (b) 50 % (2 in 4) Ff cao
- **B1** (c) 25 % (1 in 4) ff cao
- **Answer: 25 % FF (unaffected, non-carrier); 50 % Ff (unaffected carrier); 25 % ff (affected).**

Question 5

- **B1** different individuals in the population have different alleles/characteristics, oe
- **B1** if the environment changes, some individuals may already have characteristics that make them better suited to the new conditions, oe
- **B1** these individuals are more likely to survive and reproduce, passing on the advantageous alleles, so the species is more likely to survive as a whole (rather than becoming extinct), oe
- **Answer: Variation means some individuals may already suit the new conditions; they are more likely to survive, reproduce and pass on their alleles, so the species as a whole is more likely to survive rather than become extinct.**

Question 6

- **Level 3** (5-6): A complete, well-sequenced explanation of speciation via geographic isolation, correctly linking mutation, variation, differing selection pressures in the two lakes, natural selection, and eventual reproductive isolation, using accurate scientific terminology throughout.
- **Level 2** (3-4): A mostly correct explanation covering isolation and natural selection acting differently in the two lakes, but with incomplete linkage to reproductive isolation or the role of mutation/variation.
- **Level 1** (1-2): Basic statements, e.g. 'the fish changed because they were separated', with little reference to mutation, natural selection or reproductive isolation.
- **Indicative content:**
 - The geological event physically isolates the two fish populations so they can no longer interbreed (breed) with each other.
 - Mutations occur randomly in each population, producing new alleles and increasing genetic variation within each separated group.
 - The two lakes may have different conditions (e.g. different food sources, predators, water depth or temperature), so different characteristics are advantageous in each lake.
 - In each lake, natural selection favours different characteristics: individuals best suited to that lake's conditions are more likely to survive and reproduce, passing on their alleles.
 - Because the two populations cannot interbreed and are exposed to different selection pressures, the allele frequencies in the two populations gradually diverge (become more different) over many generations.
 - Over enough time (thousands of generations), the genetic differences become so great that even if the fish were reunited they could no longer interbreed to produce fertile offspring, meaning two separate species have formed (speciation).

Question 7

- **B1** (a) species A and species B are most closely related.
- **B1** (b) species D is most distantly related to species A.
- **Answer: (a) Species A and B are most closely related. (b) Species D is most distantly related to species A.**

Question 8

- **B1** evidence from chemical/molecular analysis, in particular comparing the base sequences of genetic material (DNA/RNA), or
- **B1** this evidence let scientists compare organisms at the molecular level, revealing relationships between organisms that look very different, or separating organisms that look similar but are genetically very different, or
- **B1** this is more objective/precise than comparing visible features alone, because organisms can evolve similar features independently (or physical features can be affected by environment), or
- **Answer: Comparing DNA/RNA base sequences let scientists see genetic relationships that are not visible from physical features alone, revealing that some organisms once grouped together are genetically very different.**

Question 9

- **M1** calculates before: $(40 / 500) \times 100 = 8 \%$
- **M1** calculates after: $(210 / 500) \times 100 = 42 \%$
- **A1** (b) increase = $42 - 8 = 34$ percentage points
- **B1** (c) heavy antibiotic use killed non-resistant bacteria, leaving more resources for resistant bacteria to survive and reproduce
- **B1** (c) resistant bacteria pass on the resistance allele to their offspring, so the proportion of resistant bacteria in the population increases over successive generations, or
- **Answer: Before: 8 %; after: 42 %; an increase of 34 percentage points. Heavy antibiotic use selectively killed non-resistant bacteria, so resistant bacteria survived, reproduced and passed on the resistance allele, increasing its frequency in the population.**

Question 10

- **B1** the gene for human insulin is identified and cut out of human DNA (isolated), usually using restriction enzymes
- **B1** a vector, such as a bacterial plasmid, is cut open using the same restriction enzyme so the cut ends are complementary
- **B1** the insulin gene is inserted into the plasmid and joined using an enzyme called ligase, forming recombinant DNA
- **B1** the recombinant plasmid is inserted into a bacterium, which then multiplies (replicates) and expresses the gene to produce (large quantities of) human insulin
- **Answer: Isolate the human insulin gene using restriction enzymes; cut open a plasmid vector with the same enzyme; insert and join the gene into the plasmid using ligase; insert the recombinant plasmid into bacteria, which multiply and produce human insulin.**

Question 11

- **B1** selective breeding reduces the gene pool / genetic variation of the population, because only individuals with the desired allele(s) are bred, oe
- **B1** over many generations this can lead to inbreeding, where closely related individuals are bred together
- **B1** reduced genetic variation makes the population more vulnerable to a new disease or environmental change, because there may be no individuals with alleles that give resistance/tolerance, so the whole population could be affected, oe
- **Answer: Selective breeding reduces genetic variation because only individuals with the desired traits are bred, which can lead to inbreeding. With less variation, if a new disease or environmental change occurs there may be no resistant individuals, putting the whole population at risk.**

Question 12

- **B1** the change in the DNA base sequence can change the sequence of amino acids that is coded for, oe
- **B1** this changes the way the protein (enzyme) folds, because the sequence of amino acids determines the protein's 3D shape
- **B1** if the shape of the active site changes, the substrate may no longer fit (bind), so the enzyme can no longer catalyse the reaction, oe
- **Answer: A base change can change the amino acid sequence, which changes how the protein folds. If the active site's shape changes, the substrate no longer fits, so the enzyme stops working.**

Question 13

- **B1** before meiosis begins, the DNA is copied, so each chromosome consists of two identical strands (sister chromatids)
- **B1** during meiosis, chromosomes are randomly distributed (independent assortment) into the four daughter cells, so each gamete receives a different mixture of the mother's and father's chromosomes
- **B1** two divisions occur, halving the chromosome number, so each of the four gametes produced is genetically different from the others and from the original parent cell, oe
- **Answer: DNA is copied before meiosis, then chromosomes are randomly distributed between the four gametes produced (independent assortment) across two cell divisions, so each gamete gets a different genetic mixture and the chromosome number is halved.**

Question 14

- **M1** substitutes: 92 - 45
- **A1** 47 percentage points
- **B1** as pollution decreases, tree bark becomes lighter, so dark moths are more visible to predators and are eaten more often, oe
- **B1** light-coloured moths are now better camouflaged, so they survive and reproduce more, passing on the allele for light colour, so the light allele becomes more common (dark allele becomes less common) over generations, oe
- **Answer: A decrease of 47 percentage points. As pollution fell, bark became lighter, so dark moths were more visible and eaten more, while light moths were better camouflaged, survived and reproduced more, making the light-colour allele more common.**