# FREE SAMPLE: THE FIRST QUESTION

## Question One (12 marks)

(a) Phenylketonuria (PKU) (human) is controlled by one gene with a dominant allele (P) for no PKU and a recessive allele (p) for PKU.

A man and a woman with genotypes Pp and pp have children.

(i) Complete a Punnett square and state the genotypes of the offspring.

(ii) Calculate the probability that an offspring will have PKU.

(iii) Explain the difference between the genotype and the phenotype of an individual, using one of the offspring as an example.

*[6 marks]*

(b) Phenylketonuria (PKU) is caused by a recessive allele (p). A man and a woman both have the dominant phenotype (no PKU). Their first boy child has PKU.

(i) State the genotypes of both parents and of the affected child, and explain how you worked them out.

(ii) Calculate the probability that their next child will have PKU.

(iii) The next child does not have PKU. Calculate the probability that this child is a carrier (heterozygous) and explain your answer.

*[6 marks]*

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## THE WORKED ANSWER TO THAT QUESTION

### Question 1 (12 marks) [Inheriting a characteristic]

(a) Phenylketonuria (PKU) (human) is controlled by one gene with a dominant allele (P) for no PKU and a recessive allele (p) for PKU. A man and a woman with genotypes Pp and pp have children. (i) Complete a Punnett square and state the genotypes of the offspring. (ii) Calculate the probability that an offspring will have PKU. (iii) Explain the difference between the genotype and the phenotype of an individual, using one of the offspring as an example.

**(a) Answer: (i) Pp × 2, pp × 2; (ii) $\frac{1}{2}$; (iii) explained**

- Evidence level: Achieved (gametes from each parent, Punnett square, then count).
- (i) Gametes: P and p from the first parent and p and p from the second parent. Offspring: Pp × 2, pp × 2.
- (ii) P(PKU) = $\frac{1}{2}$ (the offspring that are pp).
- Achievement: (iii) States that genotype is the alleles and phenotype is the appearance.
- Merit: Links to an example: an offspring Pp has the dominant phenotype (no PKU) but carries a recessive allele.
- Excellence: Full explanation: the genotype is the combination of alleles an individual has (for example Pp); the phenotype is the observable characteristic that results from the genotype and the environment (no PKU); because P is dominant, the genotypes PP and Pp both give the phenotype no PKU, while only pp shows PKU.

> **KEY TAKEAWAY**
> Crosses: list the gametes, complete the Punnett square, and count the offspring; probability = number of squares ÷ 4; two carriers have a 1 in 4 chance of an affected child.

(b) Phenylketonuria (PKU) is caused by a recessive allele (p). A man and a woman both have the dominant phenotype (no PKU). Their first boy child has PKU. (i) State the genotypes of both parents and of the affected child, and explain how you worked them out. (ii) Calculate the probability that their next child will have PKU. (iii) The next child does not have PKU. Calculate the probability that this child is a carrier (heterozygous) and explain your answer.

**(b) Answer: (i) both parents Pp, child pp; (ii) $\frac{1}{4}$; (iii) $\frac{2}{3}$**

- Evidence level: Merit (an affected child must have two recessive alleles, one from each parent).
- (i) The child has pp (two recessive alleles, one from each parent). The parents have the dominant phenotype, so each has at least one P, and both must carry p, so both are heterozygous: Pp.
- (ii) Cross Pp × Pp gives PP, Pp, Pp and pp in equal numbers, so P(PKU) = $\frac{1}{4}$.
- Achievement: (iii) States that the probability is $\frac{2}{3}$.
- Merit: Links to the Punnett square: the child is one of the three offspring types with the dominant phenotype (PP, Pp, Pp).
- Excellence: Full explanation: since the child does not have PKU, the child cannot be pp; of the three remaining equally likely outcomes (PP, Pp, Pp) two are heterozygous, so P(carrier) = $\frac{2}{3}$.

> **KEY TAKEAWAY**
> Pedigree reasoning: an affected child of unaffected parents shows that both parents are carriers; the probability of another affected child is $\frac{1}{4}$, and an unaffected child is a carrier with probability $\frac{2}{3}$.

*LOS: Inheriting a characteristic (92022)*

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