

15. Genetics - Terminology and Frequency Predictions

General Objectives

- Describe the interaction between genetic and environmental factors that determine the phenotype of an individual
- Discuss the ways in which genotype affects phenotype for both monogenic and polygenic traits

Content

Genetic and environmental influences on individuals. Sex determination. Autosomal, X-linked, dominant, recessive, multiple alleles, co-dominance and polygenic modes of inheritance. Examples including Huntington disease, PKU, Duchenne muscular dystrophy, and skin colour (dihybrid crosses not required). Also ABO blood groups and the existence of other blood grouping systems.

1. 1999 / 2

When two individuals, both heterozygous for a single-gene character, are crossed, all the offspring happen to resemble their parents for that character. This genetic trait is referred to as

- genotypic.
- dominance.
- phenotypic.
- recessiveness.

2. 1999 / 4

In humans, a male baby results when

- the egg is fertilised by a sperm carrying a Y chromosome.
- the egg is fertilised very early in the menstrual cycle.
- the unfertilised egg contains a Y chromosome.
- the woman's vaginal fluid is very acid (low pH).

3. 2000 / 18

A dominant gene controls tongue rolling. A man who can roll his tongue is married to a woman who cannot. They have four children, two who can roll their tongue and two who cannot. If 'A' represents the gene for tongue rolling and 'a' the recessive allele, what is the genotype of the parents?

- Man Aa x woman AA
- Man Aa x woman Aa
- Man AA x woman aa
- Man Aa x woman aa

4. 2000 / 19

Red-green colour-blindness is an X-linked recessive disorder. A mother with this condition will pass this allele to

- her daughters only.
- all of her children.
- her sons only.
- none of her children.

5. 2000 / 21

Cystic fibrosis is an autosomal recessive disease. A married couple are both carriers for this disease. If they have two children, what is the probability that **both** will be born free from this disease?

- 9/16
- 1/16
- 2/3
- 6/8

6. 2000 / 22

Which of the following is **NOT** a characteristic of an X-linked recessive disorder?

- (a) For a female to show the trait, her father must also have it.
- (b) More males than females are affected.
- (c) The trait often skips a generation from grandmother to grand-daughter.
- (d) If a woman has the trait, all of her sons will have it.

7. 2001 / 24

In a mating between a man with genotype Dd and a woman with genotype dd , the expected proportion of homozygotes among their offspring is

- (a) 25 percent
- (b) 50 percent
- (c) 75 percent
- (d) 100 percent

8. 2001 / 25

Mutations to genes on the sex chromosomes are likely to be expressed

- (a) equally in males and females.
- (b) more frequently in females.
- (c) more frequently in males.
- (d) not at all, because they are invariably lethal.

9. 2001 / 26

An allele is best defined as

- (a) a lethal recessive phenotype.
- (b) a lethal dominant phenotype.
- (c) a type of gene only found on one sex chromosome.
- (d) an alternative form of a gene at a given locus.

10. 2001 / 27

Albinism is inherited as an autosomal recessive trait. If a person has albinism and is married to a person who is heterozygous for the trait

- (a) there is a 50% chance of having a child with albinism.
- (b) there is a 25% chance of having a child with albinism.
- (c) there is a 75% chance of having a child with albinism.
- (d) there is no chance of having an albino child.

11. 2001 / 28

The genotype of an individual

- (a) is always the same as the phenotype.
- (b) refers to the visible traits of the individual.
- (c) refers to the composition of the genes.
- (d) is indicated by the presence of either an X or Y chromosome.

12. 2001 / 34

Siblings in a family may show marked inherent variations in skin and hair colour because

- (a) they differ in the amount of time they spend in the sun.
- (b) these factors are determined polygenically.
- (c) dark colouration is a dominant trait.
- (d) skin colour is related to blood group.

13. 1996 / 48

Question 13 refers to the list of words below.

LIST OF WORDS

autosome
gene
allele
chromosome
gamete
trait
karyotype
phenotype
genotype
X linked
heterozygous
homozygous

Using the most appropriate word from the list above complete **EACH** of the following definitions.

- _____ = an alternative form of a gene.
- _____ = the genetic makeup of an individual for a particular trait.
- _____ = having two different alleles for a given trait.
- _____ = the outward appearance of an organism.
- _____ = chromosome not involved in sex determination. (5)

14. 1999 / 42

NB Several of these terms would better fit different chapters but are included here to keep the question in its original form.

LIST OF WORDS

Bipedalism
Cline
Genotype
Karyotype
Homozygous
Homologous
Monogenic
Palaeontology
Pedigree
Phenotype
Polygenic
Population
Prehensibility
Stratigraphy
Superposition

Using the most appropriate word from the list above, complete **EACH** of the following definitions.

(1999 / 42 cont)

- = possessing the same pair of alleles for a given characteristic.
= study of the sequence of rock layers as a means of relative dating.
= walking upright on two legs
= the type of inheritance where the phenotype is dependent on many pairs of alleles.
= the appearance of an individual as determined by genetic constitution.

15. 1999 / 43

- (a) A healthy couple had nine children. Of these, two boys developed haemophilia (a sex-linked character resulting in an inability to form blood clots following injury) while the remaining three boys and four girls had normal blood clotting capacity.

- (i) Using "h" for the gene for haemophilia and "H" for the gene for normal blood clotting, correctly write the **mother's** genotype in relation to haemophilia. (1)
- (ii) What percentage of her four healthy daughters would be likely to be carriers of the disease? (1)

(1)

- (iii) If a haemophilic son married into a family with NO history of haemophilia, what can you predict about the likely genotype of their first-born daughter? (1)

(1)

- (iv) If a haemophilic son married into a family with NO history of haemophilia, what can you predict about the likely genotype of their firstborn son? (1)

(1)

16. 2000 / 50

- (a) A married couple has blood types **A** and **B** respectively.
- (i) In the space provided below, show how you would work out the different possible blood genotype(s) their children might inherit. (4)

(2000 / 50 cont)

- (ii) Other than the blood phenotypes of the parents, name the other possible blood phenotypes the children may inherit.

(2)

- (b) Blood typing can be used in paternity suits when the identity of the father is questioned. Briefly, explain why a blood test can only suggest who is **not** the father.

(2)

- (c) A married couple have three children who have free ear lobes, an autosomal dominant trait, and one child that has attached ear lobes. Using **F** to represent the allele for free lobes and **f** for attached lobes, what are the most likely genotypes of the parents? (1)

(1)

EXTENDED ANSWERS

17. 1997 / 51

- (a) Characteristics like haemophilia are said to be sex-linked. Explain how sex-linked characteristics are inherited. (6)

(6)

18. 1999 / 50

- Malaria is a parasitic disease widespread in the tropics and transmitted by mosquitoes. Sickle-cell anaemia is a recessive gene disorder found in a relatively high proportion (up to 40%) of people from tropical Africa and from countries around the Western Indian Ocean. People carrying a single copy of the gene, (Aa), are generally healthy and are less susceptible to malaria than people who do not carry the gene (AA). People who are homozygous for this gene (aa) are also less susceptible to malaria, but develop a form of anaemia that is almost always fatal.

- (b) Imagine you are a genetic counsellor dealing with a couple in one of the countries above. A couple wanting to have a family presents for advice. Both are known to be heterozygous for the sickle cell gene. Calculate the percentages of the likely genotypes of their children, and describe the phenotypes and survival prospects of these possible offspring. (10)

(10)