# Chapter 19 – Heredity

Subject content

**Content**

- The Passage of Information from Parent to Offspring
- The Nature of Genes and Alleles, and their Role in Determining the Phenotype
- Monohybrid Crosses
- Variation
- Natural and Artificial Selection

**Learning outcomes**

(a) define a gene as a unit of inheritance and distinguish clearly between the terms gene and allele
(b) explain the terms dominant, recessive, codominant, homozygous, heterozygous, phenotype and genotype
(c) predict the results of simple crosses with expected ratios of 3:1 and 1:1, using the terms homozygous, heterozygous, $F_1$ generation and $F_2$ generation
(d) explain why observed ratios often differ from expected ratios, especially when there are small numbers of progeny
(e) use genetic diagrams to solve problems involving monohybrid inheritance (genetic diagrams involving autosomal linkage or epistasis are not required)
(f) explain co-dominance and multiple alleles with reference to the inheritance of the ABO blood group phenotypes (A, B, AB and O) and the gene alleles ($I^A$, $I^B$ and $I^O$)
(g) describe the determination of sex in humans – XX and XY chromosomes
(h) describe mutation as a change in the structure of a gene such as in sickle cell anaemia, or in the chromosome number, such as the 47 chromosomes in the condition known as Down syndrome
(i) name radiation and chemicals as factors which may increase the rate of mutation
(j) describe the difference between continuous and discontinuous variation and give examples of each
(k) state that variation and competition lead to differential survival of, and reproduction by, those organisms best fitted to the environment
(l) give examples of environmental factors that act as forces of natural selection

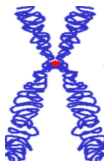
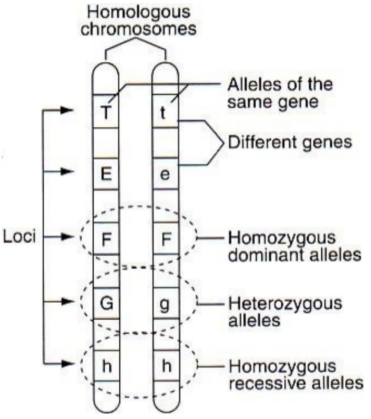
*Use the knowledge gained in this section in new situations or to solve related problems.*

Definition

| Phrase | Definition |
|---|---|
| **Chromosome** | Rod-like structure visible in nucleus during cell division<br>Made up of DNA wound around proteins.<br>Contain genetic information passed on from one generation to the next |
| **Gene** | A unit of inheritance borne on particular gene loci of chromosome<br>Segment of DNA containing sequence of nucleotides that codes for formation of polypeptide / protein that controls a characteristic / trait |
| **Homologous chromosomes** | A pair of chromosomes similar in shape and length.<br>Exactly same order of gene loci |

| | |
|---|---|
| | One chromosome in the pair come from male parent, the other come from female parent |
| **Gene loci** | Locations / positions of genes on chromosome |
| **Allele** | Different form of same gene<br>Occupy same relative positions on a pair of homologous chromosomes |
| **Pure-bred** | Individuals who are homozygous at a particular gene locus |
| **Selfing** | Fusion of nuclei of female + male gamete from same individual |
| **Test cross** | Conducted to determine genotype of organism with dominant trait by crossing it with an organism which is homozygous recessive at the gene locus in question |

## 19.1 Key Definitions

<table>
<tr><th>Term</th><th>Definition</th><th>Figure</th></tr>
<tr><td>Chromosome</td><td>Rod-like structure visible in nucleus during cell division<br>made up DNA molecules<br>carry many genes along its length</td><td>  </td></tr>
<tr><td>Gene</td><td>Unit of inheritance borne on a particular gene locus on a chromosome</td><td></td></tr>
<tr><td>Allele</td><td><u>Different forms of same gene</u> that occupy <u>same relative location</u> on a pair of homologous chromosomes
<table>
<tr><th>Type</th><th>Expressed</th></tr>
<tr><td>Dominant</td><td>homozygous & heterozygous</td></tr>
<tr><td>Recessive</td><td>homozygous</td></tr>
<tr><td>Co-dominant</td><td>2 alleles fully expressed</td></tr>
</table></td><td> 
</td></tr>
<tr><td>Phenotype</td><td>Expressed trait in an organism<br>Result of organism’s genes and effects of environment</td><td></td></tr>
<tr><td>Genotype</td><td>Genetic make-up of organism (combination of genes in organism)
<table>
<tr><th>Type</th><th>2 alleles</th></tr>
<tr><td>Homozygous</td><td>identical</td></tr>
<tr><td>Heterozygous</td><td>different</td></tr>
</table></td><td></td></tr>
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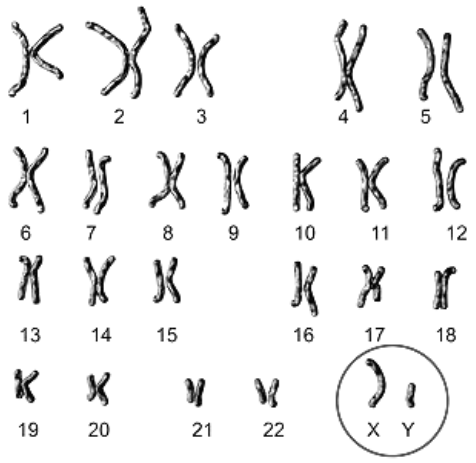
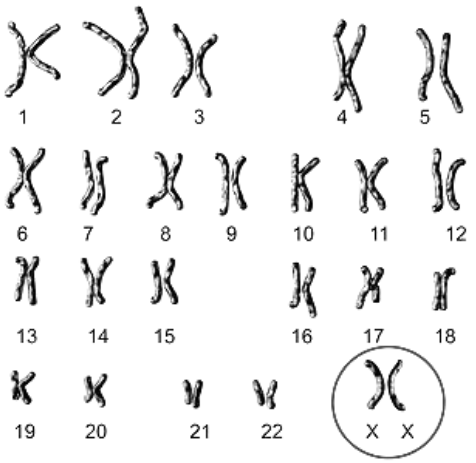
## Karyogram

| Male | Female |
|---|---|
|  |  |





Non HC: sex chromosome in male

| X chromosome | Y chromosome |
|---|---|
| longer | shorter |
| Carry genes for certain X-linked traits<br>(e.g. colour-blindness, haemophilia) | Carry fewer genes<br>(e.g. form testes, make sperms) |

## Genetic diagram

| | |
|---|---|
| **T**: tall plant<br>**t**: dwarf plant<br><br>Parental phenotype<br><br>Parental genotype<br><br>Gametes<br><br>Offspring genotype<br><br>Offspring phenotype<br><br>Offspring phenotypic ratio | |

Note:

- Must include **key** to define what each alphabet means.
  - Choose alphabet that dominant trait begins with
  - Use capital letter for dominant allele + small letter for recessive allele
- Use '**X**' when crossing
- All **arrows** must have ARROWHEADS to show direction
- **Gametes** must be CIRCLES
- Encouraged to box up all **genotypes** to avoid confusion
- **Phenotype** must be an observed trait, i.e. 'Tall plant' instead of 'tall', 'purple flower' instead of 'purple', 'round seed' instead of 'round'

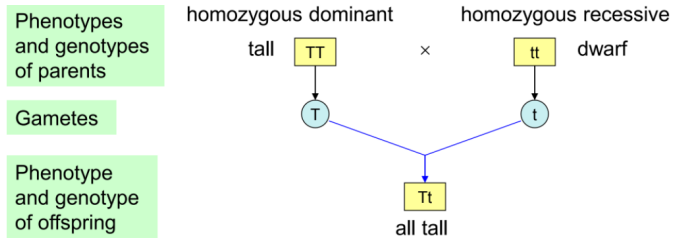
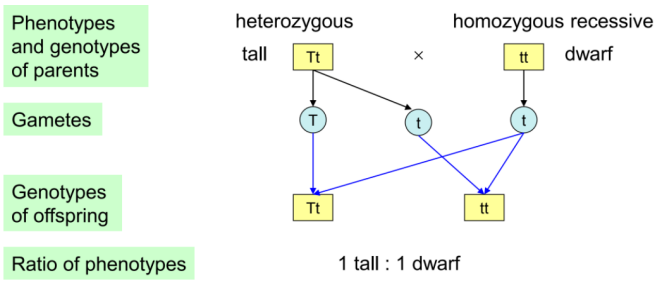
**Punnett square**

| Gametes | T | t |
|---|---|---|
| T | TT | Tt |
| t | Tt | tt |

## 19.2 Determining Genotypes

**Test cross**

Unknown organism that express dominant trait X organism that express recessive trait

| Possibility 1: **homozygous dominant** | Possibility 2: **heterozygous** |
|---|---|
| 100% offspring show dominant trait | 50% offspring show dominant trait<br>50% offspring show recessive trait |
|  |  |

**Co-dominance**

**Co-dominance**: 2 alleles controlling a trait **both express** themselves

| Example | Description |
|---|---|
| short-horned cattle | Homozygous red bull **X** homozygous white cow<br>● offspring: roan (mixture of red + white)<br>● both allele express themselves equally in hybrid → intermediate phenotype |
| four o'clock plants | Homozygous red flower **X** homozygous white flower<br>● offspring: pink flower<br>● both allele express themselves equally in hybrid → intermediate phenotype |

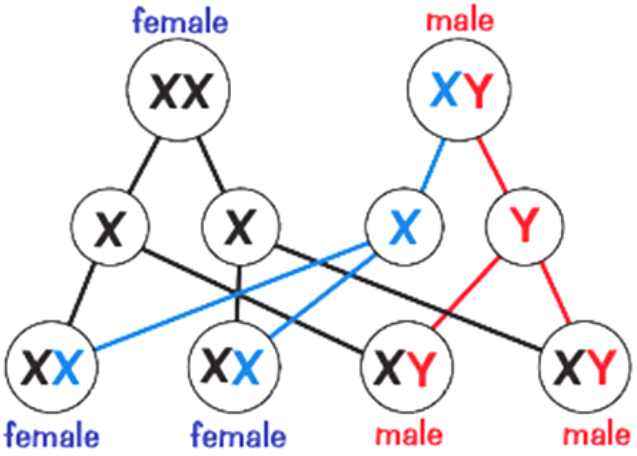
(Represent co-dominant alleles as both capital letters, e.g. $C^W$, $C^R$)

**Sex determination**

**Sex choromosomes** : chromosomes that determine sex in organism
**Autosomes** : rest of chromosomes

Human: 1 pair of sex chromosomes

- Female : **XX**
- Male : **XY**




1 female : 1 male

## 19.3 Multiple Alleles

**Multiple alleles**: gene exists in more than 2 alleles

**Human blood group**

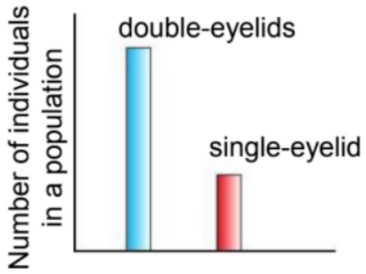
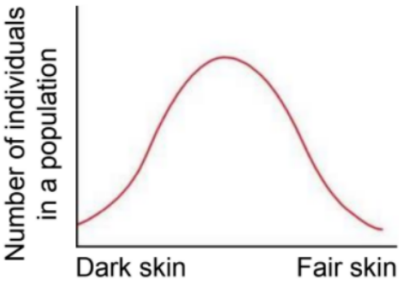
| $I^A = I^B > I^O$ |
|---|
| $I^A$ and $I^B$ are dominant over $I^O$<br>$I^A$ and $I^B$ are co-dominant |

Genotypes for blood groups

| Allele | $I^A$ | $I^B$ | $I^O$ |
|---|---|---|---|
| $I^A$ | **$I^AI^A$**<br>A | **$I^AI^B$**<br>AB | **$I^AI^O$**<br>A |
| $I^B$ | **$I^AI^B$**<br>AB | **$I^BI^B$**<br>B | **$I^BI^O$**<br>B |
| $I^O$ | **$I^AI^O$**<br>A | **$I^BI^O$**<br>B | **$I^OI^O$**<br>O |

| Blood group | Genotype |
|---|---|
| A | $I^AI^A$ or $I^AI^O$ |
| B | $I^BI^B$ or $I^BI^O$ |
| AB | $I^AI^B$ |
| O | $I^OI^O$ |

## 19.4 Continuous and Discontinuous Variations

| **Discontinuous variation** | **Continuous variation** |
|---|---|
| Deal with a few clear-cut phenotypes | Deals with a range of phenotypes |
| Controlled by one or a few genes | Controlled by many genes |
| Genes do not show additive effect | Genes show additive effect |
| Not affected by environmental conditions | Affected by environmental conditions |
| Examples<br>● ability to roll tongue<br>● gender<br>● blood group | Examples<br>● height<br>● skin colour<br>● intelligence |
|  |  |

## 19.5 Mutation

**Mutation**

**Mutation**: error during replication of gene / chromosome

- Inherited by offspring if occur in cells that give rise to gamete
- Dominant mutation: easily detected
  Recessive mutation: not detected for generations
- Types
  1. **Gene mutation** → change in structure of gene
  2. **Chromosome mutation** → change in no. of chromosome

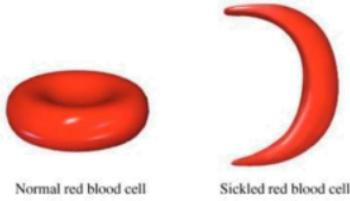
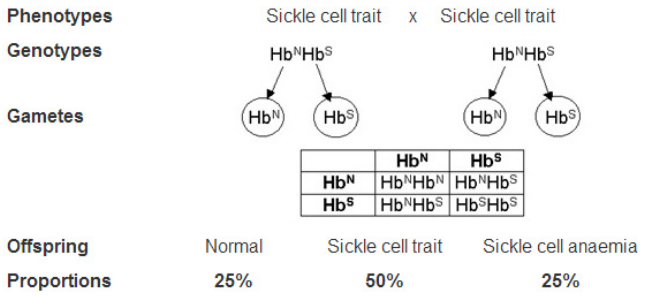
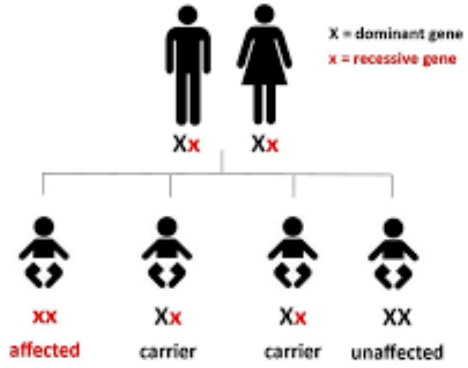
Types of mutation

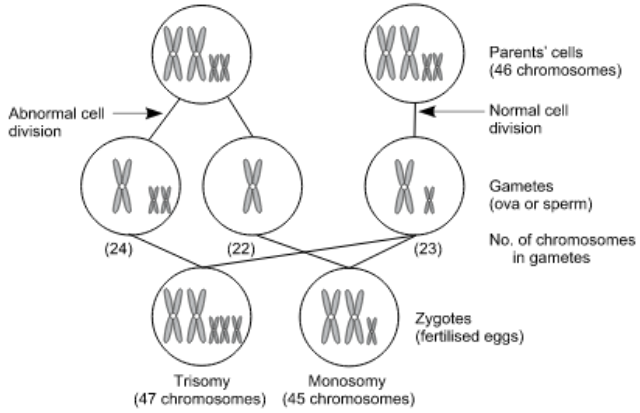
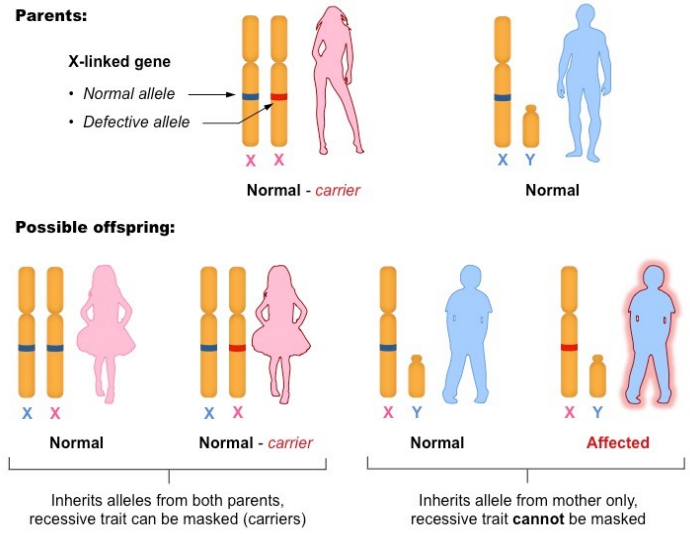
| Gene mutation | Chromosome mutation |
|---|---|
| change in structure of DNA / arrangement of nucleotides → affect sequence of amino acids → affect protein synthesised → new allele of genes | change in structure / no. of chromosomes |
| • albinism<br>• sickle-cell anaemia | • Down's syndrome |

**Mutagens** / mutagenic agent: increase rate of mutation (higher than spontaneous mutation)

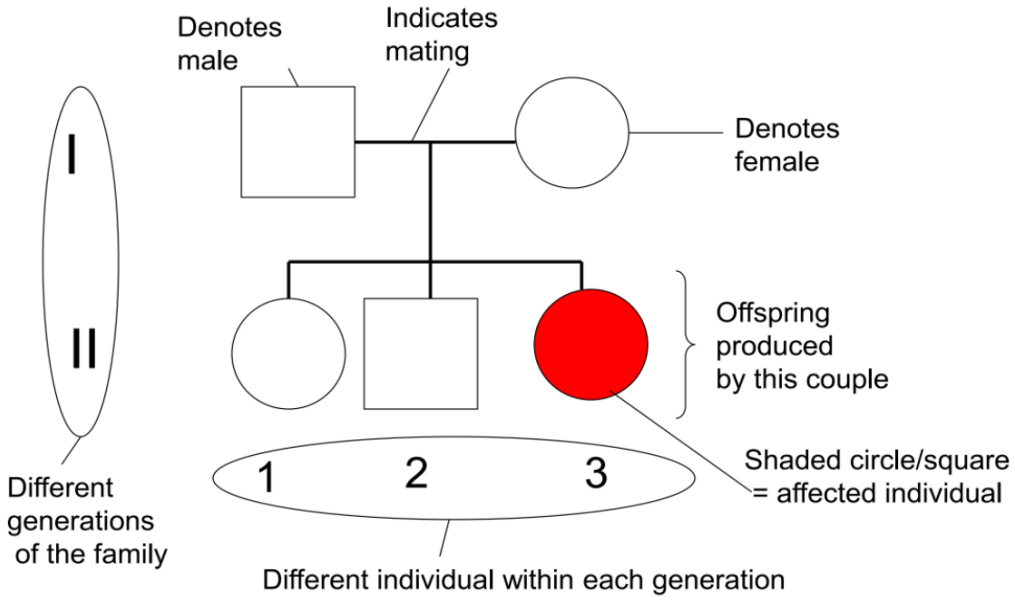
| Radiation | Chemicals |
|---|---|
| • UV light, X-ray<br>• $\alpha$, $\beta$ radiation<br>• $\gamma$ rays | • mustard gas<br>• tar<br>• formaldehyde<br>• lysergic acid diethylamide (LSD) |

Mutation

| Mutation | Description | Figure |
|---|---|---|
| 1. **Sickle cell anaemia** (gene mutation) | • Gene controlling haemoglobin production mutate → produce <u>haemoglobin S (HbS)</u><br>  ○ different in 1 <u>amino acid</u> from <u>normal haemoglobin (HbA)</u><br>  ○ change in 3D shape of haemoglobin molecule → clump together → sickle-shaped cell<br>• **Sickle-shaped RBC** (not biconcave)<br>  ○ lower ability to carry oxygen → X transport oxygen at fast rate<br>  ○ fatigue, short of breath<br><br>**Heterozygote advantage**: heterozygote (carriers) > homozygote in malaria-infected countries<br>• Malaria: infect RBC containing HbA → rupture → lower no. of RBC<br>• HbS: resistance to malaria (unaffected) → survive |  <br>  |
| 2. **Albinism** (gene mutation) | • Gene mutation caused by <u>recessive allele</u> → <u>absence of pigment</u> in skin, hair, eyes<br>• **Albino**: people who suffer from albinism<br>  ○ reddish-white skin<br>  ○ white hair (no melanin)<br>  ○ red iris (blood vessels) |  |

| | | |
|---|---|---|
| 3. **Down's syndrome** (chromosome mutation) | • Error in cell division → <u>extra copy of chromosome 21</u><br>  ○ Pair of chromosome 21 in sperm / ovum X separate<br>  ○ Embryo develops: extra chromosome is replicated in every cell<br>• Effects<br>  ○ reduced life span<br>  ○ mental retardation<br>  ○ heart defects<br><br>**Trisomy**: extra copy of chromosome<br>**Monosomy**: only one copy of chromosome |  |
| 4. **Edward's syndrome** (chromosome mutation) | • Error in cell division → <u>extra copy of chromosome 18</u><br>• Effects<br>  ○ defects in brain, heart, stomach, kidneys<br>  ○ die before birth |  <br>18 |
| 5. **Colour blindness** (gene mutation) | • Changes in DNA sequence on X chromosome<br>• Sex-linked genetic disorder<br>• Effects<br>  ○ difficulty seeing / differentiating colours | Inheritance of X-linked recessive disease<br>  |

**Pedigree tree**




## 19.6 Selection

**Natural selection**

**Natural selection**

- Best adapted organisms survive → reproduce → pass on genes to next generation
- Nature selects varieties that are:
  - more competitive
  - more resistant to diseases
  - better adapted to environmental changes
- **Evolution**: simpler ancestral → present complex forms of living organisms

Mechanism of evolution:

1. Mutation in indv in species → new alleles → variation of traits within species
2. Compete with each other over limited resources
3. Varieties with favourable trait have selective advantage
4. Varieties better adapted to changes in envt → more likely to survive + reproduce → pass on beneficial alleles to offspring
5. Over many generations: beneficial alleles accumulate in varieties → different from original stock (most of population made out of indv with the trait) → replace original stock
6. Give rise to new species

**Artificial selection**

**Artificial selection**

- Selective breeding: animals and plants with favourable inheritable traits artificially selected + breed → improved varieties
- Purpose:
  1. Higher yields of better quality
  2. Increased resistance to pests and diseases
  3. Increased tolerance to extreme weather and temperatures (e.g. drought, frost)

**Hybridisation**: breeding b/w different varieties to produce offspring with combined desirable traits

Mechanism:

| Breeding within animal / plant variety | Hybridisation |
|---|---|
| 1. Select indv with desirable traits + cross<br>2. From progeny: select those that express desirable traits to highest degree + cross<br>3. Continue inbreeding to fix desirable genes within variety of breed<br>● produce indv homozygous for desirable genes<br>● ensure that future generations inherit desirable genes | 1. Select indv with diff desirable traits + cross<br>2. From progeny: select those that express combinations of desirable traits + cross<br>3. Continue inbreeding to fix desirable genes |

Comparison

| Aspect | **Natural selection** | **Artificial selection** |
|---|---|---|
| 1. Cause | Varieties produced by mutations | Varieties produced by selective breeding |
| 2. Selection force | Environment / Nature | Man |
| 3. Speed | Very slow | Relatively fast |
| 4. Adv to Man | May be advantageous / harmful | Advantageous |
| 5. Parents | X consciously chosen | Consciously chosen for desirable traits |

## Typical questions

### Multiple choice questions

**1** Which two statements about continuous variations are correct? (N2011/P1/Q38)

1. The heights of adult humans will partly depend on the quality of their diets when young.
2. The faster period of growth in humans is the embryo.
3. A group of adult males had heights ranging from 155 cm to 220 cm.
4. Humans have stopped growing by the time they are 22 years old.
5. Humans grow taller during babyhood and childhood.

**A** 1 and 2
**B** 1 and 3
**C** 2 and 4
**D** 3 and 5

**2** The diagram shows six male and six female birds of the same species.

females males

Which row describes the type of variation shown in these birds?

(N2015/P1/Q38 / N2019/P1/Q38)

| | pattern of spots on the wings of the female birds | presence or absence of ring around the eye | sizes of males and females |
|---|---|---|---|
| **A** | continuous | discontinuous | continuous |
| **B** | continuous | discontinuous | discontinuous |
| **C** | discontinuous | continuous | continuous |
| **D** | discontinuous | continuous | discontinuous |

**3** These statements describe some genetic terms.

1. inheriting the same allele from both parents
2. inheriting different alleles from each parent
3. the genetic composition of an organism
4. the observable characteristics of an organism

Which row contains the correct genetic terms for the descriptions? (N2019/P1/Q36)

| | 1 | 2 | 3 | 4 |
|---|---|---|---|---|
| **A** | heterozygous | homozygous | homologous | genotype |
| **B** | homologous | codominant | phenotype | genotype |
| **C** | homozygous | codominant | homologous | phenotype |
| **D** | homozygous | heterozygous | genotype | phenotype |

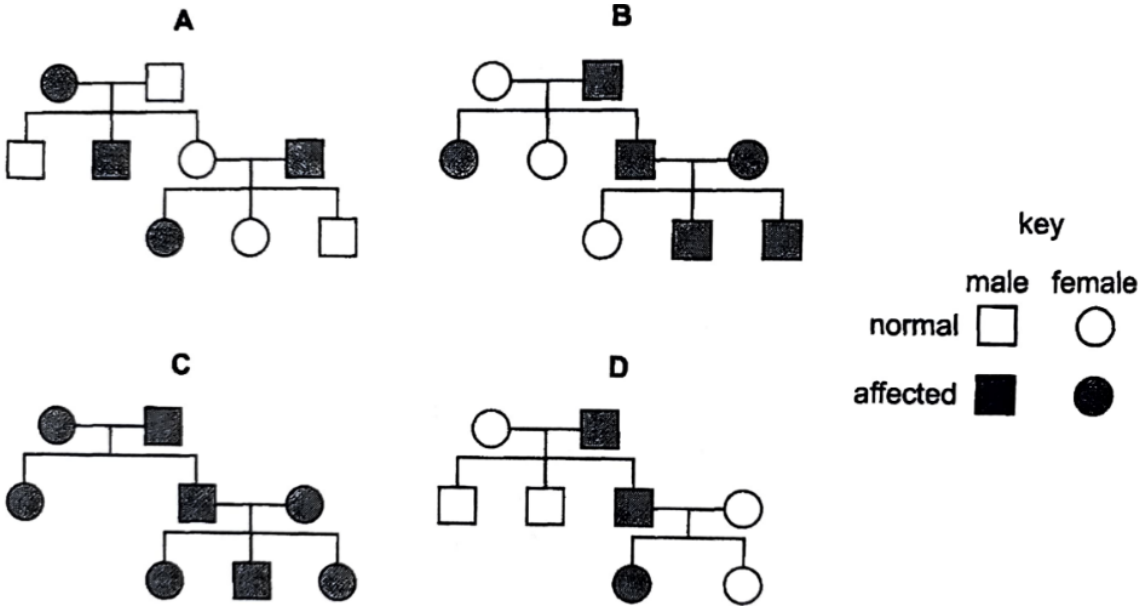
**4** A scientist studies the length of beaks in a population of birds for a number of generations. The scientist concluded that the length of the beaks was an example of continuous variation. Which information about the length of beaks supports this conclusion? (N2019/P1/Q37)

**A** The beaks were longer in males than in females.
**B** The length of the beak depended on the type of seed eaten.
**C** Long beaks were pale in colour; short beaks were dark in colour.
**D** The length of the beaks had a range with no distinct categories.

**5** In terms of natural selection, what is the significance of the survival of the fittest in a tiger population? (N2011/P1/Q36)

**A** They achieve the best physical condition.
**B** They contribute the most offspring to the next generation.
**C** They have the most effective camouflage.
**D** They are the most sexually active.

**6** The inheritance pattern of an abnormal condition in four families is shown.
Which family proves that the condition must be caused by a dominant allele?
(N2011/P1/Q37 / N2018/P1/Q38)



**7** Changes in the climate may lead to the melting of sea ice and the thawing of ice on the land in the Antarctic that has been frozen for a very long time.
What could lead to evolution in this situation? (N2012/P1/Q38)

**A** Animals adapt their features to suit the new environment.
**B** Previously advantageous features may become disadvantageous.
**C** Seeds dormant for thousands of years could germinate.
**D** There would be less competition for space to live.

**8** In mice, the allele for black fur colour is dominant to the allele for white fur colour.
What does this mean in a mouse population? (N2013/P1/Q36)

**A** Mice with black fur are more successful breeders.
**B** Most mice have black fur.
**C** When a black-furred mouse breeds with a white-furred one, the offspring will have grey fur.
**D** White-furred mice are only born to two white-furred parents.

**9** A brown-eyed man and a brown-eyed woman have five children, three with brown eyes and two with blue eyes.
Which statement about this family is correct? (N2014/P1/Q37)

**A** Both blue-eyed children have the same genotype and phenotype.
**B** The alleles for brown eyes and blue eyes are co-dominant.
**C** The genotypes of the parents must be the same as the genotypes of the three brown-eyed children.
**D** The three brown-eyed children must be heterozygous for eye colour.

**10** Which statement uses genetic technology and information correctly? (N2014/P1/Q38)

**A** Recessive alleles are usually harmful and dominant alleles are beneficial.
**B** When two animals are crossed, one homozygous dominant for fur colour and the other homozygous recessive, their offspring will each have different phenotypes.
**C** When two people, both homozygous for blood group A, have children, both parents and offspring will have the same genotype.
**D** Under the ABO blood grouping system, alleles $I^A$, $I^B$ and $I^O$ are co-dominant.

**11** In an investigation, two mice are mated.
The $F_1$ generation all have the same fur colour. The $F_1$ generation is then crossed with each other to produce an $F_2$ generation where there is a 3:1 ratio for fur colour.
What states the genotypes for fur colour of the **original** two mice? (N2015/P1/Q36)

**A** Both parents are heterozygous.
**B** Both parents are homozygous dominant.
**C** One parent is homozygous recessive and the other is heterozygous.
**D** One parent is homozygous dominant and the other is homozygous recessive.

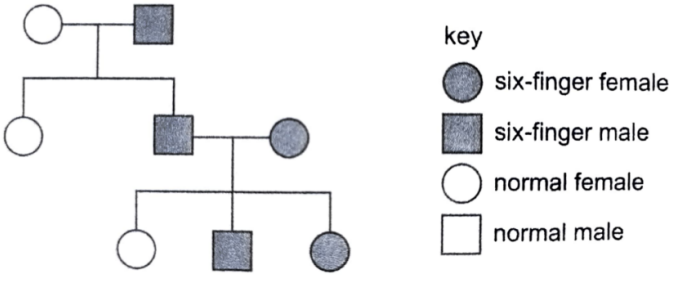
**12** Four children from different families are tested to identify their blood group. The blood group phenotypes of the four children are A, AB, B and O.
The blood group phenotypes of the four sets of parents are shown in the table.
Which couple are the parents of the child with blood group AB? (N2016/P1/Q37)

| | mother | father |
|---|---|---|
| **A** | A | B |
| **B** | B | B |
| **C** | O | AB |
| **D** | O | O |

**13** Bacteria evolve resistance to the antibiotic penicillin by the process of natural selection.
Which factor acts as the force for this natural selection? (N2016/P1/Q38)

**A** competition with other bacteria
**B** ultraviolet radiation
**C** use of antibiotics
**D** use of disinfectants

**14** A mutation sometimes occurs in humans which causes each hand to have six fingers.
The diagram shows how this condition is inherited in a family.




What does the family tree show about the mutated allele? (N2017/P1/Q37)

**A** It is co-dominant.
**B** It could be dominant or recessive.
**C** It is dominant.
**D** It is recessive.

**15** The statements refer to a mouse population.
The allele for black fur colour is dominant to the allele for white fur colour.

1. Mice with black fur are less successful breeders.
2. Most mice have white fur.
3. Two black-furred mice can produce white-furred offspring.
4. Two white-furred mice produce offspring all with white fur.
5. When a black-furred mouse breeds with a white-furred one, the offspring will have grey fur.

Which statements are correct? (N2018/P1/Q37)

**A** 1 and 2
**B** 2 and 5
**C** 3 and 4
**D** 4 and 5

**Structured questions**

**1** Distinguish clearly between

**(a)** gene and allele

- A gene is a heritable sequence of nucleotides along a DNA molecule, which codes for a polypeptide that determines a particular trait.
- An allele is an alternative form of a given gene.

**(b)** dominant and recessive

- A dominant allele will express its trait in a homozygous or heterozygous condition. In a heterozygous condition, the dominant allele will mask / suppress the recessive allele.
- The recessive allele is expressed only when another identical allele is present in a homozygous.

**(c)** phenotype and genotype

- Phenotype is the physical appearance of an organism.
- Genotype is the specific combination of alleles of a gene.

**2** A pure-bred tall plant was crossed with a pure-bred dwarf plant. A total of 600 offspring were produced of which all were tall. The tall offspring were selfed. A total of 2700 plants were found in the second filial ($F_2$) generation.
Show by using a genetic diagram, the ratio of tall plants to dwarf plants in the $F_2$ generation. Using the results of your cross, predict the number of dwarf plants in the $F_2$ generation.

**Genetic Diagram:**
T: allele for tall trait
t: allele for dwarf trait

| | | | | |
|---|---|---|---|---|
| parental phenotype | tall plant | | dwarf plant | |
| parental genotype | TT | X | tt | |
| gametes | T | | t | |
| $F_1$ generation genotype | | **Tt** | | |
| $F_1$ generation phenotype | | All tall | | |
| selfing of $F_1$ generation | Tt | X | Tt | |
| gametes | T | t | T | t |
| $F_2$ generation genotype | **TT** | **Tt** | **Tt** | **tt** |
| $F_2$ generation phenotype | tall | tall | tall | dwarf |
| $F_2$ phenotypic ratio | | 3 tall : 1 dwarf | | |

$\therefore$ No. of tall $F_2$ generation plants = 2025
No. of dwarf $F_2$ generation plants = 675

**3** Two short haired guinea pigs are mated several times and produced 100 offspring of which 27 have long hair.
What are the probable genotypes of the parents? Verify your answer with the help of a Punnett square.

H: allele for short hair
h: allele for long hair

**Working:**
Probable genotype of the guinea pigs: Hh X Hh

**Punnett Square:**

| Gametes | H | h |
|---|---|---|
| H | **HH** | **Hh** |
| h | **Hh** | **hh** |

Since the phenotypic ratio of the cross is = 73 short hair : 27 long hair
= 2.7 short hair : 1 long hair
which is close to the phenotypic ratio of 3:1 when 2 heterozygous parents are crossed, as shown in the Punnett square above.

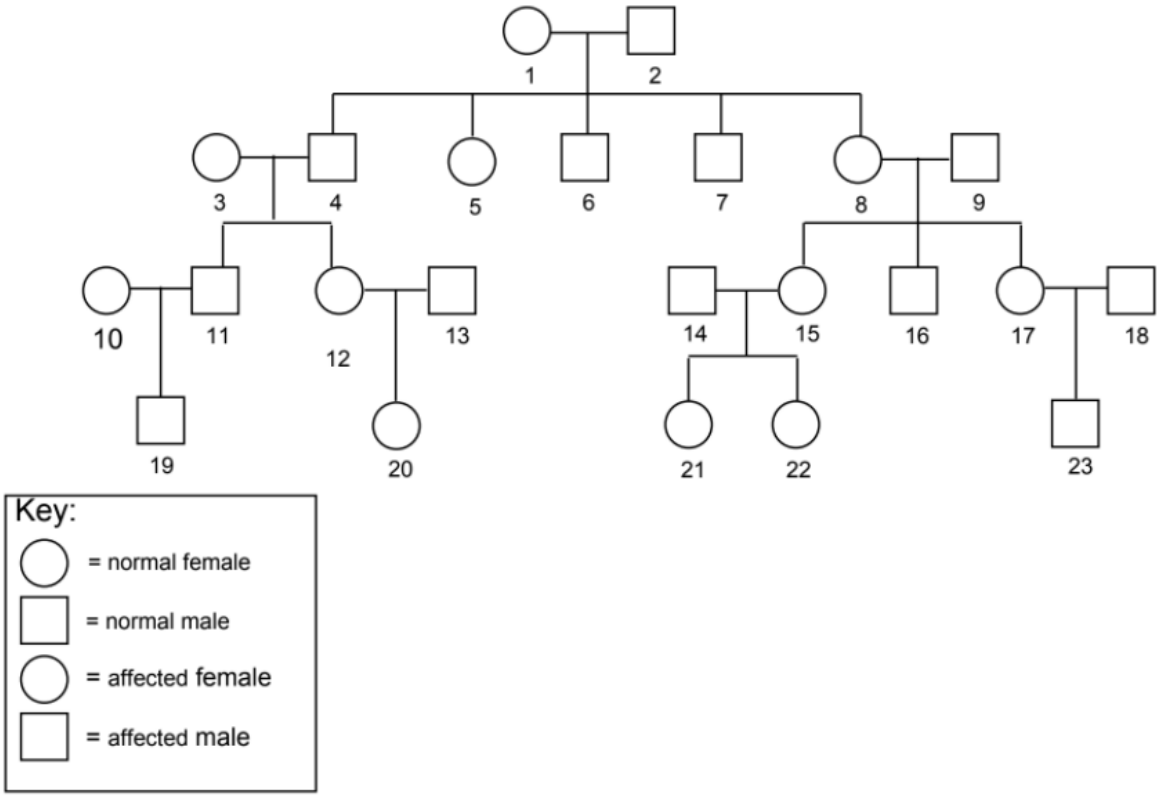
**4** What would a test cross show if the genotype of the organism in question is heterozygous?

- If the individual is heterozygous, then half his offspring will express the dominant trait and the remaining half will express the recessive trait.
- If all the offspring in this cross express the dominant trait, the parental genotype is homozygous.

**5** In sweet pea plants, **Y** is the dominant allele for yellow pods and **y** is the recessive allele for green pods. A farmer wishes to determine the genotype of a sweet pea plant that has yellow pods. He did a test cross and got a phenotypic ratio of 1:1 in the offspring.
Explain what is the unknown genotype of the sweet pea plant the farmer is interested in. (Genetic diagram and Punnett square are not allowed in your answers)

- Since half the offspring have yellow pods and the other half have the green pods, the sweet pea plant is heterozygous.
- Presence of green pea pods in the offspring means that the green pea pod offspring must have inherited one recessive allele from the yellow parent plant. Hence, the yellow pea pod plant must be heterozygous.

**6** Huntington's chorea (HC) is a neurological disorder marked by muscular twitching which appears around 35 to 45 years of age and progresses to the point of total incapacitation and finally death over a period of 15 years. Huntington's chorea is inherited as an autosomal dominant. Below is a pedigree of a family affected by Huntington's chorea.




**(a)** Which individual, if any, is homozygous for the trait?

None. If any of the individual is homozygous dominant for the trait, they would have inherited one dominant HC allele from each parent. Hence, both parents should be affected with HC. None of the affected individuals in the diagram has both parents that are also affected by HC.

**(b)** Individual 11 died at age 26 in a car accident. Describe and explain the chances of his son being affected.

As the disorder does not show up until 35 to 45 years of age, there is a 50% chance that individual 11 might develop the condition.

H: allele for Huntington's chorea
h: allele for normal condition

| gametes | H | h |
|---|---|---|
| h | Hh | hh |
| h | Hh | hh |

Based on the Punnett square, if individual 11 has the disease, there is a 1⁄2 chance that the son is affected by the disease.

Therefore, the chance of his son being affected
= 1⁄2 X 1⁄2
= 1⁄4

**(c)** Comment on the chances of individual 23 having the trait.

The probability is zero as neither grandparent (8 and 9), expected to be well over 45 years by now, show any signs of the disease and thus would not have carried the dominant Huntington allele.

**(d)** Draw a genetic diagram to show how individuals 1 and 2 pass the condition to their children.

| | | | | |
|---|---|---|---|---|
| Parent | **1** | | **2** | |
| Parental phenotype | Normal | | Huntington's chorea | |
| Parental genotype | hh | | Hh | |
| Gametes | h | h | H | h |
| Offspring genotype | Hh | Hh | hh | hh |
| Offspring phenotype | 1 Huntington's chorea | | 1 normal | |

**(e)** Is Huntington's chorea an example of continuous variation or discontinuous variation? Explain your answer.

Discontinuous variation: an individual either show the symptoms or not at all

**(f)** Why is it that such a deleterious gene as that for Huntington's chorea is not selected against by natural selection?

Natural selection can only act by affecting reproductive potential or affecting the number actually reproducing. Since Huntington chorea strikes in the middle years after reproductive age, neither the reproduction potential or rate are affected. Affected individuals are still able to pass on the dominant HC allele to their offspring.

**7** Explain how an albino child may be born to normal parents.

- Albinism is a recessive trait caused by recessive alleles (genotype: aa)
- Normal parents of the albino child are heterozygotes (carriers) with the genotype Aa.
- Their child would have inherited one recessive allele (a) for albinism from each parent for the albinism phenotype to be expressed.

**8** Explain how mutation, variation and natural selection can lead to the formation of a new species of living organism.

1) Despite high reproductive rate, number of organisms in a population of a species remains fairly constant due to environmental factors, competition for resources and predation.
2) Mutation give rise to new alleles/variation + some alleles/variations are advantageous
3) When competition exists in the environment, nature selects individuals with advantageous / favourable traits such as being more competitive, disease-resistant or adaptable to changes in the environment.
4) Organism with advantageous variation survives to reproductive age to pass on the beneficial gene to offspring.
5) As a result, the frequency of the new gene or allele will increase in the population.
6) Organisms with new traits may arise after many years, giving rise to a new species.

**9** Using named examples, explain what is meant by a mutation. [4]
(N2017/P2/B10)

A mutation is a sudden modification of the structure of a gene or in the chromosome number.

- For example, sickle cell anaemia in humans is caused by a gene mutation. The gene controlling haemoglobin production is mutated and produces abnormal haemoglobin which clumps together. The red blood cells are sickle-shaped as a result and cannot transport oxygen efficiently.
- Down's syndrome is an example of chromosome mutation. Normal humans have 46 chromosomes. However, people with Down's syndrome have 47 chromosomes because they received one more chromosome 21 during fertilisation. They have characteristic facial features and face mental and physical difficulties.

**10** (N2014/P2/A7)

**(a)** State the three alleles of the gene that controls human blood groups. [1]

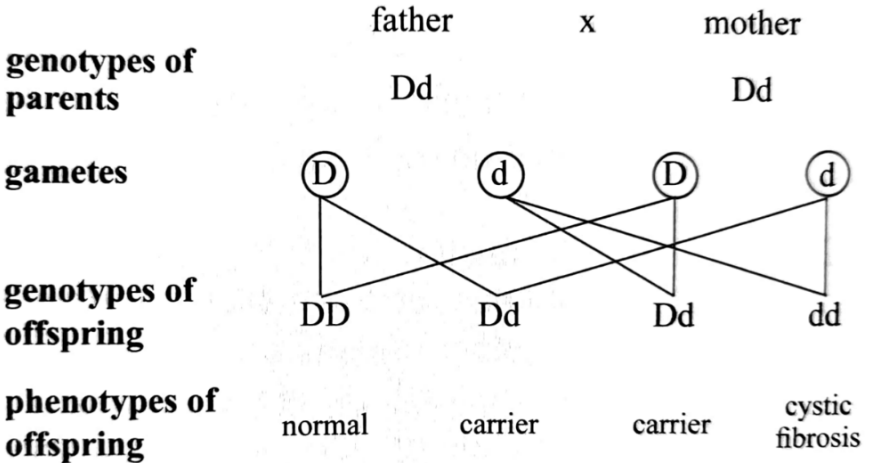
$I^A$, $I^B$, $I^O$

**(b)** Complete the genetic diagram below to show the possible blood groups of the children of a father who is heterozygous for blood group A and a mother who is heterozygous for blood group B. [5]

| | father | | mother | |
|---|---|---|---|---|
| genotypes of parents | $I^AI^O$ | | $I^BI^O$ | |
| gametes | $I^A$ | $I^O$ | $I^B$ | $I^O$ |
| genotypes of offspring | $I^AI^B$ | $I^AI^O$ | $I^BI^O$ | $I^OI^O$ |
| phenotypes of offspring | AB | A | B | O |

**11** (N2019/P2/A4a,b)

**(a)** A father and mother are both carriers (heterozygous) of the allele for the disease cystic fibrosis. Cystic fibrosis is caused by a recessive allele.
Complete the genetic diagram. Use the symbol **D** for the dominant allele and **d** for the recessive allele. [4]




**(b)** A couple has a one in two chance of having a child with cystic fibrosis. One of the parents has cystic fibrosis and the other parent is a carrier for the cystic fibrosis allele.
The couple has two children. These children do not have cystic fibrosis.
Explain why the actual ratio is different from the expected ratio. [1]

As the couple has only two children,

the sample size is too small to be statistically significant.

**12** With reference to the inheritance of ABO blood group phenotypes, explain what is meant by *co-dominance*. [6]
(N2017/P2/B10 OR)

Co-dominance occurs when two alleles controlling a trait both express themselves in the phenotype. Neither allele is completely dominant over the other.

There are four different blood groups in humans. They are A, B, AB and O. The blood groups are determined by three alleles $I^A$, $I^B$, $I^O$. Each person has any two of the alleles. Under the ABO blood grouping system, alleles $I^A$ and $I^B$ are co-dominant while allele $I^O$ is recessive.

- Allele $I^A$ is dominant over allele $I^O$, so individuals with genotypes $I^AI^A$ or $I^AI^O$ have blood group A.
- Allele $I^B$ is also dominant over allele $I^O$, so individuals with genotypes $I^BI^B$ or $I^BI^O$ have blood group B.

- Allele $I^O$ is recessive, so only individuals with genotype $I^OI^O$ have blood group O. Allele IO does not express itself in the heterozygous condition. It only expresses itself in the homozygous condition.
- Both alleles $I^A$ and $I^B$ express themselves in the phenotype in both homozygous and heterozygous conditions. They are co-dominant to each other. Thus, individuals with genotype $I^AI^B$ have blood group AB.

**13** Study the pedigree tree diagram to illustrate in eye colours of Daniel's family. The allele **B**, for brown eyes is known to be dominant while the allele **b**, for blue eyes is recessive.

**(a)** State the possible genotypes for the brown eye colour and blue eye colour in this family. [2]

Bb for brown eyes
bb for blue eyes

**(b)** State why one of Daniel's children does not have any child with blue eyes. [2]

The ratio of her getting a brown-eyed child to a blue-eyed child is 1 : 1 since her husband is homozygous recessive for blue eyes.
Hence, there is a 50% chance of getting either a brown-eyed or a blue-eyed child every time she gives birth. In this case, all the three children happen to receive allele B from their mother during random fertilisation, thus they have brown eyes.

**(c)** State the type of variation that is observed in the colour of eyes. [1]

Discontinuous variation

**(d)** Suppose Daniel's great granddaughter is marrying an individual with blue eyes, determine, using a genetic cross, the percentage of a child born with brown eyes. [2]

Daniel's great granddaughter has blue eyes.

Based on the genetic cross above, it can be concluded that the percentage of a child born with brown eyes is 0% since the genotype of the offspring is all homozygous recessive.