The big picture

? Guiding question(s)

- What patterns of inheritance exist in plants and animals?
- What is the molecular basis of inheritance patterns?

Keep the guiding questions in mind as you learn the science in this subtopic. You will be ready to answer them at the end of this subtopic. The guiding questions require you to pull together your knowledge and skills from different sections, to see the bigger picture and to build your conceptual understanding.

Are you unique?

Have a look at the people around you. There are many traits that you can observe. The colour of their eyes or whether they have attached or detached earlobes are clearly visible to you. For those you are biologically related to, you might see shared traits. You might have the family 'dimple' or have the same widow's peak hairline as your father (**Figure 1**).

Credit: razyph, Getty Images

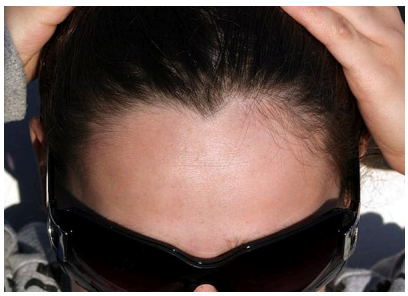
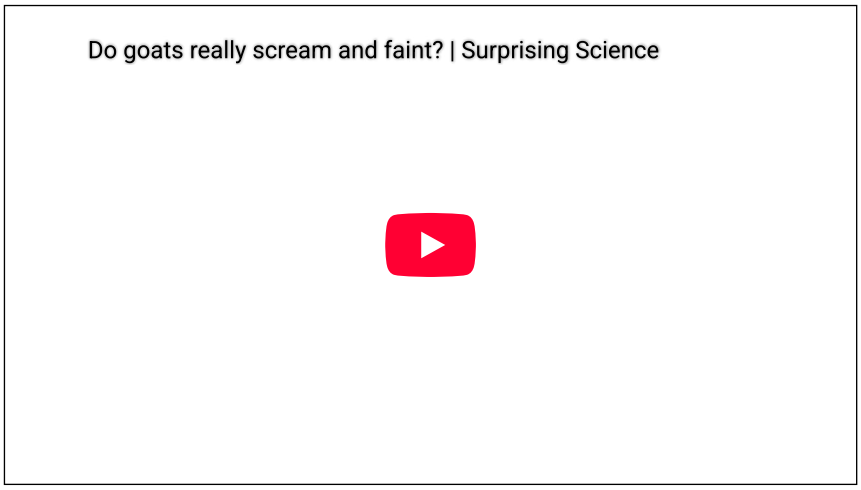


Figure 1. Physical traits like a dimple or a widow's peak are inherited.

But have you ever met a stranger that looks just like you? The use of social media has made it easier for us to study the faces of unrelated individuals. Facial recognition software algorithms allow an objective measure of 'likeness'. A study in 2022 (https://www.smithsonianmag.com/smart-news/doppelgangers-dont-just-look-alike-they-also-share-dna-180980635/) looked at 16 pairs of 'doppelgangers'. When they tested the genetic makeup of these similar individuals, researchers found that nine of the pairs shared many common genetic variations. So their likeness was more than just skin deep!

Fainting goats

There are many other traits that you cannot see, however, such as a person's ability to produce insulin or lactase, for example. Another example of a hidden genetic anomaly can be found in a breed of domestic animal called the Tennessee fainting goat. These goats appear normal, but when they get startled or excited, their muscles go stiff and they seem to faint! The goats have a condition called myotonia congenita, an autosomal dominant trait. A missense mutation caused by a single nucleotide substitution results in the amino acid proline being produced instead of alanine within the gene for a skeletal muscle chloride channel. The muscle does not get supplied with sufficient chloride ions and hence becomes stiff, resulting in the goats falling over, as seen in **Video 1**. Do all genetic changes give such drastic results or can some changes be silent?



Video 1. Tennessee fainting goats will appear to faint when startled or excited.

In the following section, you will learn about specific patterns of inheritance and how traits are passed on from one generation to the next. You will also learn how we can present this information in a graphical way.

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Prior learning

Before you study this subtopic make sure that you understand the following:

- The difference between male and female gametes (see subtopics B2.3 (/study/app/bio/sid-422-cid-755105/book/big-picture-id-43533/) and D3.1 (/study/app/bio/sid-422-cid-755105/book/the-big-picture-id-43252/)).
- How gametes are formed and how they come together to form a zygote (see subtopics D2.1 (/study/app/bio/sid-422-cid-755105/book/big-picture-id-43548/) and D3.1 (/study/app/bio/sid-422-cid-755105/book/the-big-picture-id-43252/)).
- That there are different alleles present in a population (see subtopic D4.1 (/study/app/bio/sid-422-cid-755105/book/the-big-picture-id-43238/)).

D3. Continuity and change: Organisms / D3.2 Inheritance

From haploid to diploid

D3.2.1: Fusion of haploid gametes in parents to form a diploid zygote D3.2.2: Methods for conducting genetic crosses in flowering plants D3.2.3: Genotype D3.2.4: Phenotype

Learning outcomes

By the end of this section you should be able to:

- Outline that haploid cells (with a single copy of a gene) produced by each parent can fuse to form a diploid zygote with two copies of a gene.
- Explain methods for how flowering plants are genetically crossed.
- Distinguish between genotype (combination of inherited alleles) and phenotype (observable traits resulting from genotype plus environmental factors).

Unique you!

You may have been told that you look more like your mother, but your sister has some traits similar to your father. So, why can offspring sometimes resemble one parent more than the other if the offspring are always an equal blend of both parents? We will see why this can happen during this section.

In nature, some organisms reproduce asexually, resulting in offspring that are genetically identical clones. In contrast, eukaryotic plants and animals that have a sexual life cycle share a common pattern of inheritance which results in increasing the variation in a population. The offspring produced have 50% of the genetic material from each parent. However, unless you are a monozygotic twin, your genetic makeup is specific to you and not the same as that of your sibling, even with the same parents.

Once an individual is at sexual maturity, cells within specialised organs called gonads will start to undergo a certain kind of cell division called meiosis. The gonads contain cells whose nuclei have pairs of chromosomes. As described in subtopic D2.1 (/study/app/bio/sid-422-cid-755105/book/big-picture-id-43548/), the cells divide twice during meiosis and the unique cells produced are given the term gametes. These are known as haploid (n) as they have only one copy of each chromosome from one of the parents.

Student view

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The gametes are highly specialised with specific features. Adaptations of human male and female gametes are noted in subtopic B2.3 (/study/app/bio/sid-422-cid-755105/book/big-picture-id-43533/). On their own, these gametes cannot generate a new individual. Two gametes (one from the male parent and one from the female parent) fuse together to form a diploid (2n) zygote that now contains an equal amount of genetic information from both parents.

In flowering plants, such as the pea (**Figure 1**), the male and female gametes are often present in the same plant; such plants are described as being hermaphroditic.

Credit: Diane Miller, Getty Images

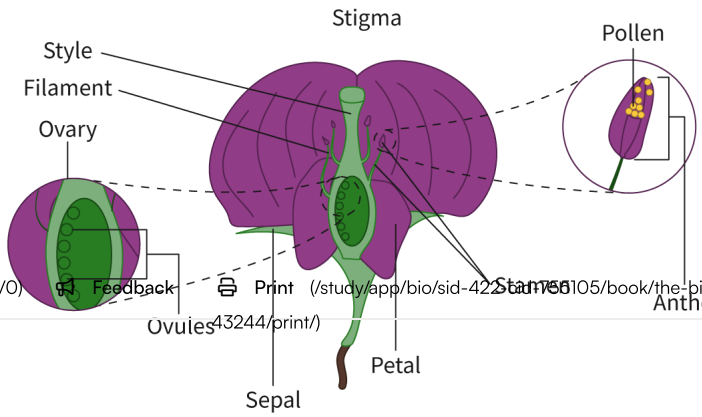

Figure 1. The flower of the garden pea plant.

More information for figure 1

This diagram depicts the flower of a garden pea plant with labeled parts. At the center, the flower's key components are highlighted, including petals and sepals that form the outer layers. The central structure includes the stigma, style, and ovary, with the filament supporting the anthers that produce pollen. To the left, a magnified view of the ovary shows the ovules inside. To the right, a close-up of the stamen highlights the anther covered in pollen. Dotted lines connect these magnified sections to their corresponding parts on the main diagram. The labels help identify each part: stigma, style, filament, ovary, and pollen among others.

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Mendel's experiments

A monk named Gregor Mendel (1822–84) from an area now found in the Czech Republic, conducted numerous plant-breeding experiments over many years using the humble pea (*Pisum sativum*). He observed flower colour along with the texture and colour of pea seeds and pods (**Figure 2**). His experiments are so important that he is often referred to as the 'father of genetics'.

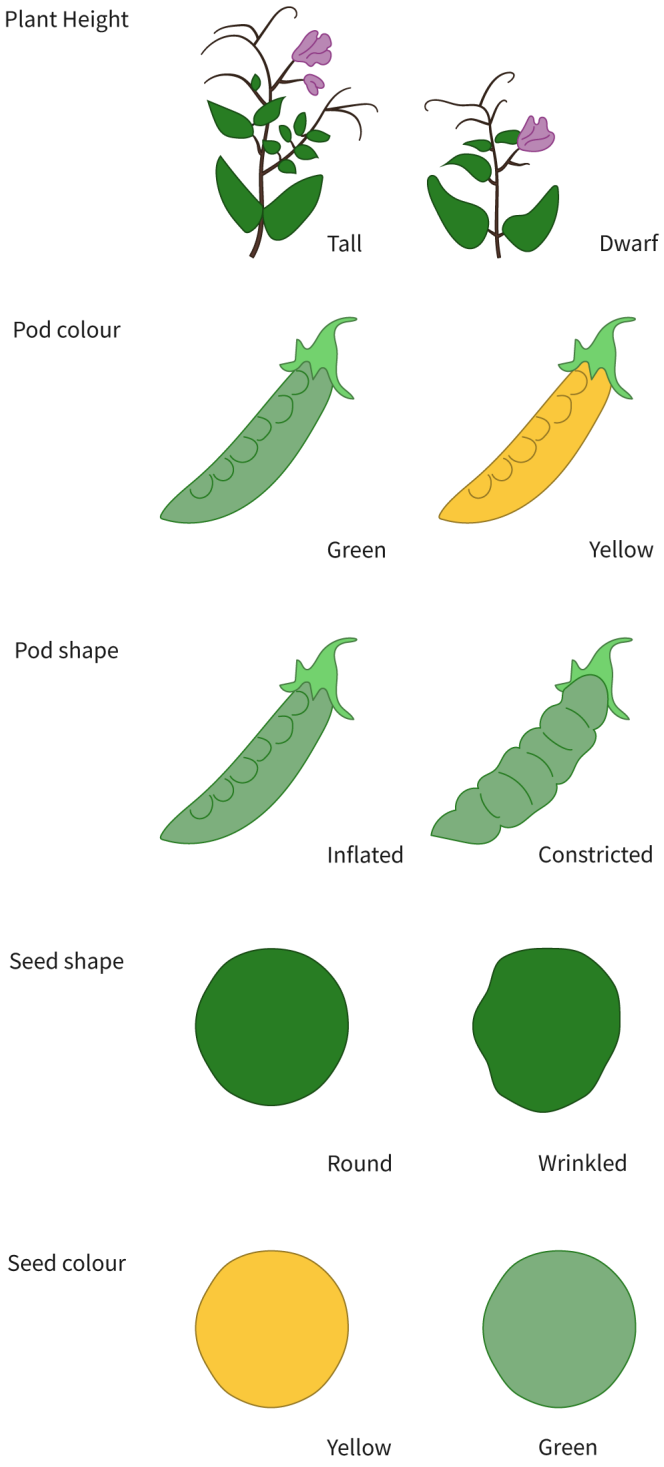

Figure 2. Mendel predicted the inheritance of various traits by observing many pea crosses.

More information for figure 2

Student view

This image is a diagram illustrating various traits of pea plants that Gregor Mendel studied in his genetic experiments. It is divided into several sections:

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1. **Plant Height:** Depicts two pea plants: one tall and one dwarf, both flowering.
2. **Pod Colour:** Shows two pods, one green and one yellow.
3. **Pod Shape:** Displays two pod shapes, one inflated and one constricted.
4. **Seed Shape:** Illustrates two seed shapes, one round and another wrinkled.
5. **Seed Colour:** Shows two seed colours, one yellow and another green.

Each pair of traits is labeled and visually distinct, representing the variations Mendel used to predict inheritance patterns. This diagram highlights the contrasting characteristics that led to Mendel's discoveries in genetics.

[Generated by AI]

To appreciate his experiments, we should understand the structure of a flower.

The male part of the flower is referred to as the stamen and comprises an elongated anther, held up by a thin filament. The anther contains the male gametes called pollen. The female part of the flower is the pistil and has a swollen base called an ovary that surrounds the female gametes known as ovules. A style extends from the ovary and terminates in a structure called the stigma. When the pollen grains are mature, the anther will break open to reveal them.

In order for the male and female gametes to come together, the pollen needs to move to the stigma. When it arrives there, it grows a pollen tube, which carries the pollen containing the haploid nucleus all the way down the hollow style to an ovule of the ovary. If pollen from one plant lands on the stigma of the same plant it is called self-pollination. If pollen from one plant lands on another plant of the same species, it is cross-pollination. Both types of pollination are illustrated in **Figure 3**. Pollen can only grow a pollen tube if it lands on the same species. However, the pollen/stigma interaction is highly selective and will try to stop pollen from the same plant growing a pollen tube, preferentially allowing pollen from a different plant of the same species. This improves the genetic variation.

Student view

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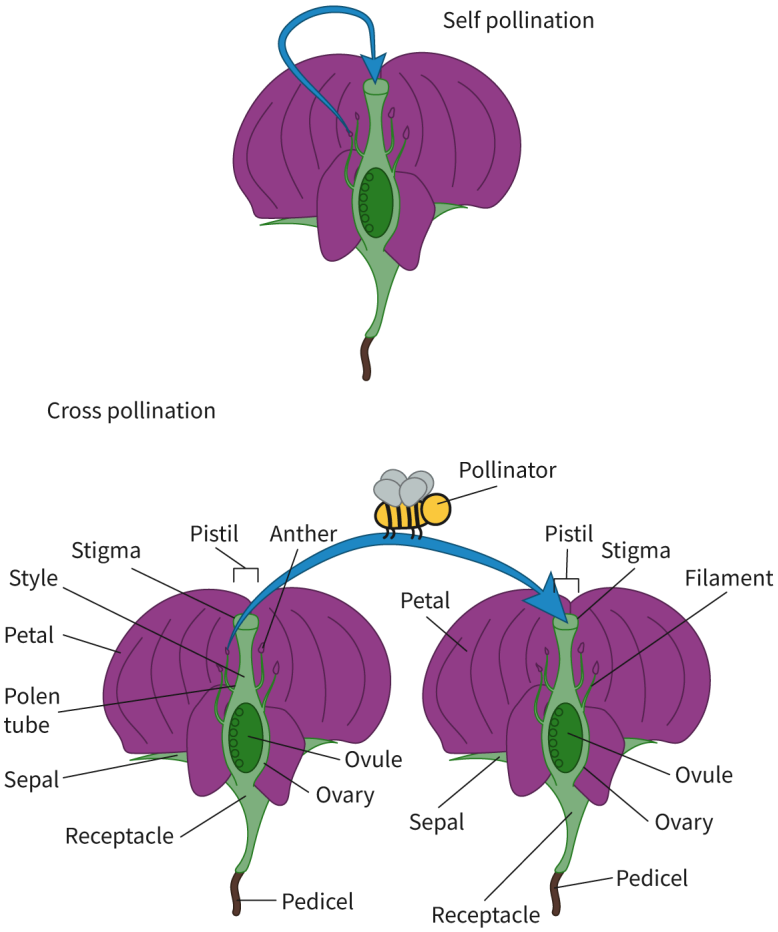

Figure 3. Self-pollination occurs between the same flower or between different flowers on the same plant. Cross-pollination occurs between different plants of the same species.

More information for figure 3

The image is an illustration of pollination types in flowering plants. At the top, there is a depiction of self-pollination. A single flower with labeled parts like "Pistil" and "Stigma" is shown with a blue arrow from the anther to stigma, indicating the movement of pollen within the same flower.

Below, the illustration shows cross-pollination. There are two flowers side by side, each with labeled parts such as "Stigma," "Pistil," "Anther," "Petal," "Sepal," and "Ovule." A bee, labeled as "Pollinator," is depicted on a blue arrow that connects the anther of one flower to the stigma of another, illustrating the process of pollen transfer between different flowers on separate plants. The image includes detailed labeling of flower parts to clarify the pollination processes.

[Generated by AI]

In his experiments, Mendel crossed many pea plants by selecting the pollen from one plant and brushing it onto the stigma of another plant. He was able to first observe and later predict the inheritance of different pea flower and pea seed traits. For example, he showed that when true-breeding parents for traits such as round seeds were bred with true-breeding parents for wrinkled seeds, they would result in offspring that produced only one form of these traits (e.g. round seeds). The parent generation is termed the P generation and the offspring are called the filial (F) generation. The first generation is therefore called the F1 generation. If two members of the F1 generation are subsequently bred together, this would give rise to the F2 generation which would then possess some physical characteristics not seen in the F1 generation (e.g. wrinkled seeds). This is illustrated in **Figure 4**.

Student view

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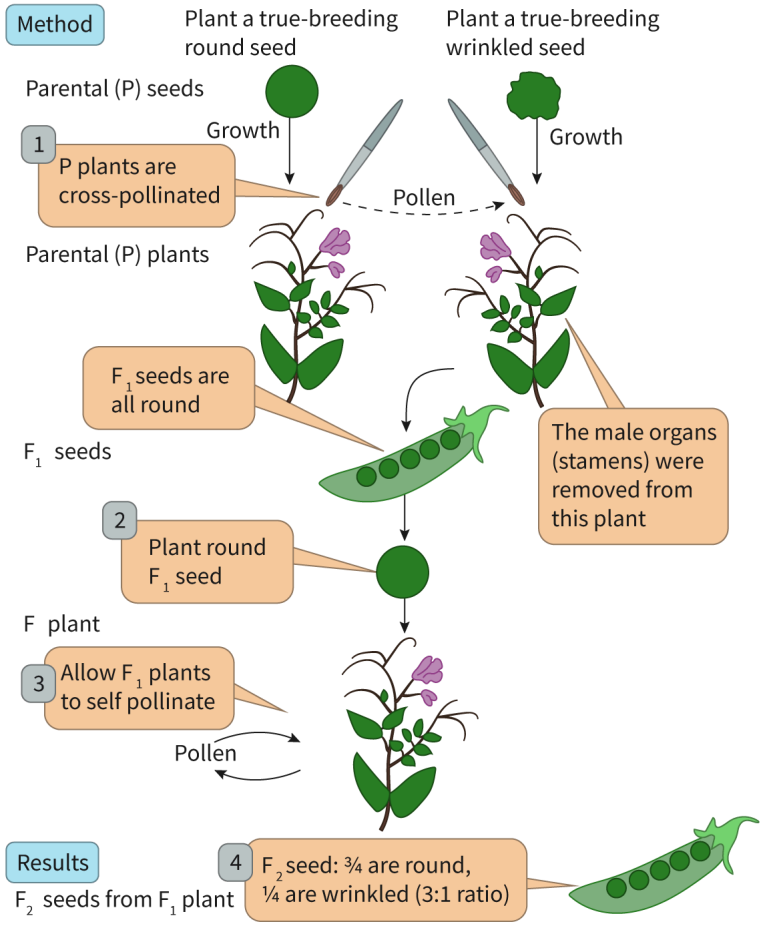

Figure 4. Mendel conducted breeding experiments with true-breeding pea plants to observe the F1 and F2 generations.

More information for figure 4

This diagram illustrates Mendel's pea plant cross-pollination experiment. It shows step-by-step methods and results. The process starts with parental (P) seeds, one true-breeding for round seeds and the other for wrinkled seeds. Both plants grow, and it's indicated that the male organs (stamens) were removed from one plant. In step 1, the P plants are cross-pollinated using a brush to transfer pollen. This results in F1 seeds which are all round, as shown in a pea pod with round seeds labeled 'F1 seeds are all round.'

In step 2, an F1 seed is planted, growing into an F1 plant. It then proceeds to step 3, where the F1 plant is allowed to self-pollinate. This is depicted with a circular motion indicating pollen transfer within the same plant.

The result in step 4 shows that the F2 generation produces seeds that display a 3:1 ratio, with ¾ being round and ¼ wrinkled, symbolized with a pea pod illustration and accompanying text 'F2 seed: ¾ are round, ¼ are wrinkled (3:1 ratio).'

[Generated by AI]

A graphical way of predicting such genetic crosses is using a Punnett grid (also called a Punnett square). These will be explained later in subtopic D3.2 (/study/app/bio/sid-422-cid-755105/book/the-big-picture-id-43244/).

Breeding climate-smart crops

Genetic breeding experiments between two related species are commonly used to generate new varieties of crops. Specific individuals can be carefully selected to ensure that the offspring have desired characteristics. As the world strives to achieve the UN Sustainable Development Goal #2 (https://www.un.org/sustainabledevelopment/hunger/) of

Student view

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Zero Hunger, scientists around the world are focused on breeding plants that are able to cope with climate change, extreme weather, drought or flooding.

International Mindedness

Sustainable development

Various academic institutions are examining how crops can be bred to be 'climate-smart'. The International Wheat and Maize Improvement Center (CIMMYT (https://www.cimmyt.org/news/historic-release-of-six-improved-wheat-varieties-in-nepal/)) has developed drought- and heat-resilient wheat that is also high in zinc. It is hoped that this will improve the situation in Nepal by providing a much-needed nutrient in a crop that can cope with climate changes.

A virus-resistant breed of potato called Unica has been developed to thrive in flood-prone and high heat areas of Kenya.

In Saudi Arabia, salt-tolerant Galapagos tomatoes have been bred with domesticated tomato plants that give nice juicy fruits to combine both these traits. The resultant plants can potentially be watered with sea water to provide a means of irrigation without the need for valuable fresh water. See **Video 1** for more information on this.

Video 1. Breeding salt tolerant crops.

These examples could all help mitigate food insecurity.

Genotypes and phenotypes

In any eukaryotic cell, pairs of homologous chromosomes exist; one chromosome comes from the mother and the other from the father. Along the length of the chromosome are stretches of DNA called genes, many of which encode specific proteins. These genes are in identical positions on each of the chromosomes and so an individual will therefore have two copies of the gene (one from each parent) (**Figure 5**).

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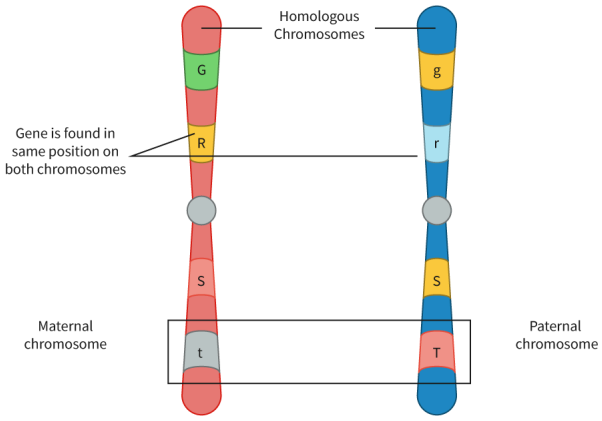

Figure 5. A pair of homologous chromosomes showing the presence of a gene in the same position on both chromosomes.

More information for figure 5

The image is a diagram depicting a pair of homologous chromosomes, each labeled as maternal and paternal. The maternal chromosome is on the left, colored in shades of red, and the paternal chromosome is on the right, colored in shades of blue. Both chromosomes have colored segments, each labeled with different letters indicating the presence of specific genes.

Starting from the top, the maternal chromosome has a green segment labeled 'G', followed by a yellow segment labeled 'R', then a red segment labeled 'S', and finally a grey segment labeled 't'. Correspondingly, the paternal chromosome features a blue segment labeled 'g' at the top, followed by a blue-yellow segment labeled 'r', then the same red segment labeled 'S', and a red segment labeled 'T' at the bottom.

The diagram also includes connecting lines, indicating that the same gene is located at identical positions on both chromosomes. This illustrates genetic concepts such as homozygosity and heterozygosity where different alleles (e.g., 'G' vs. 'g', 'R' vs. 'r') are present at the same loci on homologous chromosomes. There's also text indicating that genes can be found in the same position on both chromosomes, highlighting the concept of alleles and gene positioning on homologous chromosomes.

[Generated by AI]

However, the two genes may differ from each other by a few bases. These different forms of a gene are called alleles and this allele combination is called the genotype. When a gene has two identical alleles, this is known as homozygous. When the two alleles are different from one another, it is referred to as heterozygous. This idea is illustrated in **Figure 6**.

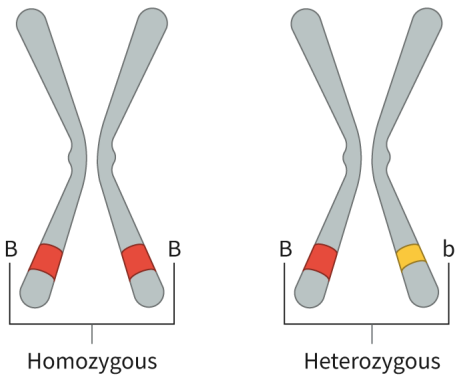

Figure 6. Different genotypes: homozygous and heterozygous.

More information for figure 6

Student view

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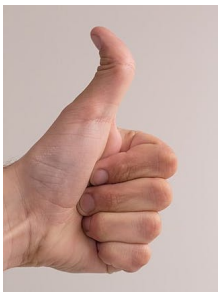
The image is a diagram illustrating the concept of genotypes using two chromosome pairs to represent homozygous and heterozygous conditions. On the left side, the chromosomes are labeled as "B" and are colored red, symbolizing a homozygous genotype where both alleles are identical. On the right side, one chromosome is labeled "B" and colored red, while the other is labeled "b" and colored yellow, indicating a heterozygous genotype where the alleles are different. Each chromosome pair is marked with respective labels below them: "Homozygous" for the left pair and "Heterozygous" for the right pair.

[Generated by AI]

Genotypes can be hard to determine but some physical traits are easily visualised and are caused by the genotype an individual possesses. For example, hitchhiker's thumb is a genetically determined condition. It is thought to be inherited in an autosomal recessive manner, meaning that a child has to inherit two copies of the gene for hitchhiker's thumb - one from each parent. However, there is still much to be learned regarding this particular trait. For example, are just two alleles involved? Is there just one form of Hitchiker's Thumb? Only time and new research will tell. In contrast, physical traits like having a tattoo are things that have been done to an individual and are not a result of the genotype. The outward expression of the combination of the alleles along with the influence of environmental factors is known as the phenotype. Some traits, like our skin colour, are a combination of both genetics and environment; in this case, how much we expose our skin to the sun. **Figure 7** illustrates the three different types of trait.



Credit: Eugenio Marongiu, Getty Images

Credit: John White Photos, Getty Images

Student view

Figure 7. Three different traits: hitchhiker's thumb is a genetic trait; having a tattoo is an environmental trait; sunburn is an example of a trait influenced by genetics and the environment.

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Try the activity below alongside **Interactive 1** to help with your understanding of parts of a flower and the processes of pollination and fertilisation.

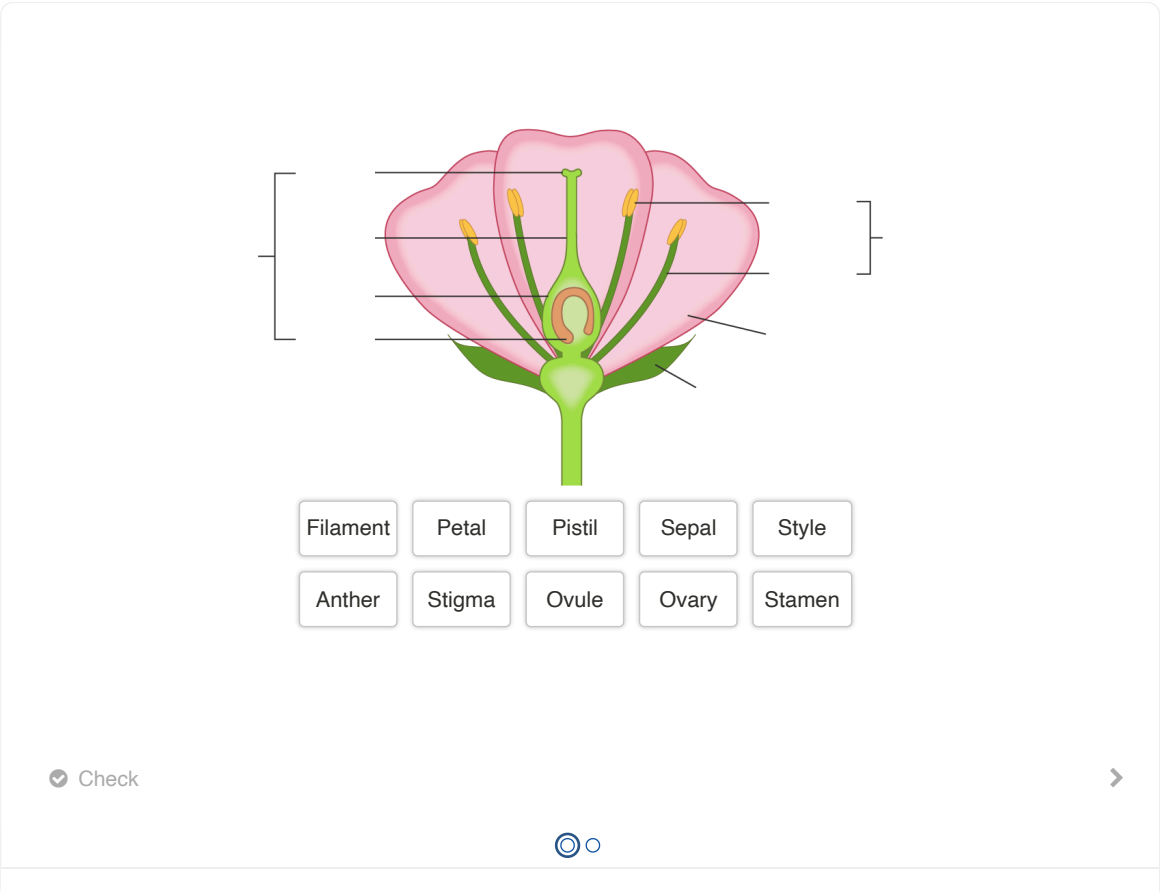
Activity

- **IB learner profile attribute:** Inquirer
- **Approaches to learning:** Research skills — Comparing, contrasting and validating information
- **Time required to complete activity:** 20 minutes
- **Activity type:** Individual activity

Your task

Step 1: In **Interactive 1** (slide 1) drag and drop the correct terms to label the insect-pollinated flower.

Step 2: In **Interactive 1** (slide 2) drag and drop the correct terms to label the parts of a flower Diploid or Haploid.



Interactive 1. Parts of a Flower.

More information for interactive 1

A two-screen interactive.
The first one is a drag-and-drop interactive, which shows an image of a simplified, vertical cutaway view of a flower. The flower is mostly pink and green and is positioned against a white background.
At the very bottom is a thick green stalk extending upwards. This stalk widens slightly into a rounded, green base from which two leaf-like green structures curve outwards on either side. Emerging from the center of the green base are several elongated, thin green stalks extending upwards. At the tip of each of these green stalks is a small, elongated, and divided yellow structure. These are arranged around a central structure.
The central structure is also green at its base and extends upwards. It swells at the bottom to form a bulbous, light green shape containing a smaller, almost oval structure in peach color. Above this swollen area, the green structure narrows into a stalk that extends

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upwards and terminates in a slightly wider and curved structure.
Three petals in pink surround the central structure and the yellow-tipped green stalks. The petals overlap each other, creating the outer form of the flower.
There are about four solid black lines on the left and right sides of the flowers ending with dotted drop boxes. The four boxes on the left are connected to a single drop box. On the other hand, the first two drop boxes on the right are connected to a single drop box.
There are 10 options given at the bottom of the flower anatomy. The labels include Filament, Petal, Pistil, Sepal, Style, Anther, Stigma, Ovule, Ovary, and Stamen.
Read below for solutions.
The labels on the left from top to bottom read: Stigma, Style, Ovary, and Ovule. The common label on the extreme left is Pistil. The labels on the right from top to bottom read: Anther, Filament, Petal, and Sepal. The common label on the extreme right is Stamen.
The right arrow at the bottom right helps to navigate to the next interactive.
The second interactive is about Haploid and Diploid cells.
On the left side, there are two rectangular boxes stacked vertically. The top box contains a light pink circle representing a cell. Inside this circle, there are two short (blue) and two long (pink), thick lines representing chromosomes. Below the circle, the caption "Diploid" is written in black text. The bottom box also contains a light pink circle representing a cell. Inside this circle, there is one short (blue), and one long (pink) thick line representing chromosomes. Below the circle, the caption "Haploid" is written in black text.
In the middle and right side of the screen, there are four drop boxes. The boxes on the top read Petal cell and Pollen and the boxes on the bottom row read Ovule and Zygote from left to right.
Read below for solutions.
Petal cell and Zygote are Diploid, and Pollen and ovule are Haploid.
The left arrow on the bottom right allows the users to navigate to the previous interactive.
The “Check” button at the bottom left allows users to check their answers for both interactives. The total number of correct answers is indicated with a slider. When wrong answers are inputted, a “Retry” button occurs at the bottom left that allows the users to redo the interactive.

Step 3: For the parts of a flower involved in pollination and fertilisation, research the number of chromosomes in these cells. Copy and complete **Table 1** with the information you find out.

Table 1. Chromosomes in cells.

Cell type	Number of copies of each chromosome	Haploid (n) or diploid (2n)
petal		
pollen		
ovule		
zygote		

Discuss your findings with the class.

5 section questions

D3. Continuity and change: Organisms / D3.2 Inheritance

Expression of phenotypes

D3.2.3: Genotype D3.2.4: Phenotype D3.2.5: Effects of dominant and recessive alleles on phenotype D3.2.6: Phenotypic plasticity D3.2.7: Phenylketonuria

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Learning outcomes

By the end of this section you should be able to:

- Contrast the effects of dominant and recessive alleles on phenotype.
- Explain how phenotypic plasticity can change across an individual's life span.
- Summarise the cause of the human disease phenylketonuria.

Genetics of tasting

You might never have thought about this, but your genes can even change the way you taste things! Let's take the case of tasting for a bitter chemical called phenylthiocarbamide (PTC). Your teacher might be able to share some paper strips that are impregnated with this chemical (**Figure 1**). If you are permitted, you can do the taste test yourself.

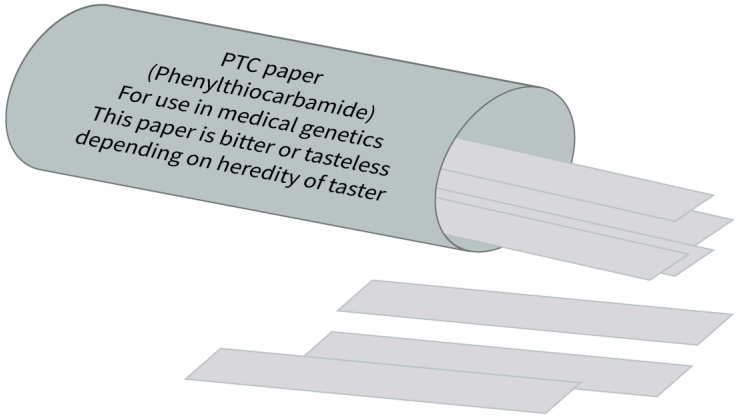


Figure 1. Some people can taste the bitter taste of phenylthiocarbamide (PTC) on these paper strips; others cannot.

More information for figure 1

The image is a diagram showing a cylindrical container labeled "PTC paper (Phenylthiocarbamide) For use in medical genetics This paper is bitter or tasteless depending on heredity of taster." The container is open at one end, with several paper strips extending out. These strips are used in genetic testing to determine the ability to taste the chemical phenylthiocarbamide (PTC). The text on the container explains that the taste perception can be either bitter or tasteless, determined by the genetic makeup of the individual.

[Generated by AI]

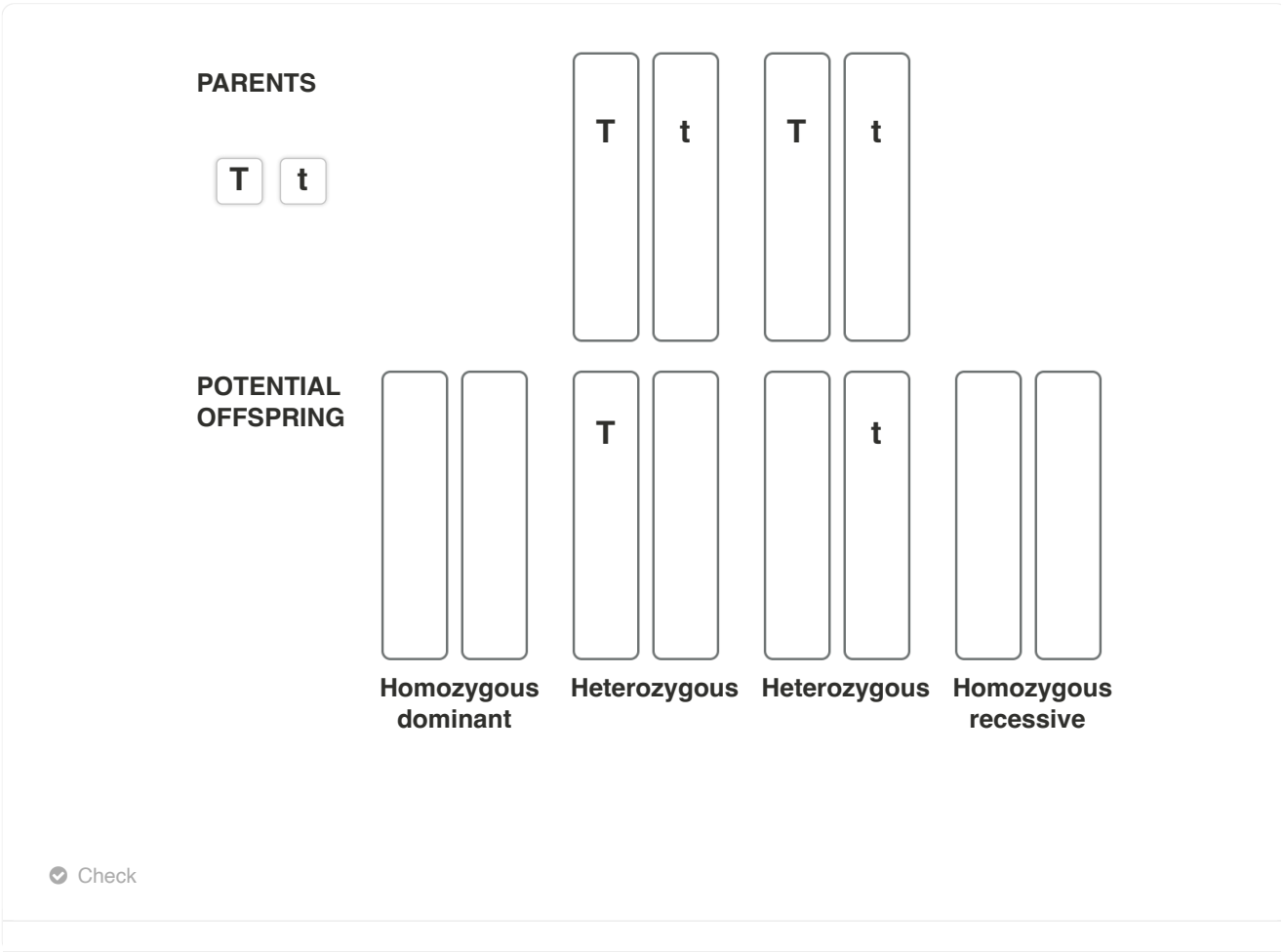
The ability to taste the bitter chemical is controlled by one gene called TAS2R38 which codes for a protein for a taste receptor on the tongue. There are two common forms of the allele: one allele is the 'tasting allele' and one the 'non-tasting' allele.

These two alleles code for a receptor protein with slightly different shapes. A person who is heterozygous, with both forms of the allele, can taste the bitter PTC chemical. The 'tasting allele' is, therefore, dominant over the 'non tasting' allele. The allele that has the effect masked is called the recessive allele.

Those that are homozygous dominant can also taste the chemical. Only those that have two 'non-tasting' alleles taste nothing at all. We say they are homozygous recessive for this allele. This is similar to the case of tongue rolling shown earlier. Only those that are homozygous recessive for the non-rolling genes cannot roll their tongue. Tongue rollers must

Student view

therefore be homozygous dominant or heterozygous for this gene. Use **Interactive 1** to generate offspring that cannot taste the chemical, as well as offspring with two different genotypes that would be able to taste the chemical.



Interactive 1. Generate all the possible genotypes the two heterozygous parents (Tt) could produce.

PTC is similar in structure to the toxic compounds found in some poisonous plants. So, the ability to taste this bitter chemical may be important in making us averse to bitter and toxic compounds and is a way that has evolved to keep us safe. Thankfully though, PTC is not poisonous!

Phenotypic plasticity

So far, we have discussed how the phenotype is dictated by an individual's genotype. However, in nature, the phenotype observed may be more complicated than this. The phenotype can be shaped by a combination of the genotype *plus* the particular environment an organism is exposed to. This ability to be shaped or moulded is called plasticity, and so this phenomenon is called phenotypic plasticity. It allows individuals with the same genome to adapt when exposed to different environmental conditions. These changes may involve an alteration in behaviour, physiology or morphology. This is not caused by changes in the genotype and these changes do not even need to be permanent; the changes might be reversed during the individual's lifetime depending on the situation. Phenotypic plasticity is important as a means to increase the chances of survival in a changing global climate. An example of this phenotypic plasticity is the seasonal polyphenism of the butterfly *Bicyclus anynana* (**Figure 2**).

Overview (/study/ap 422-cid-755105/o

Credit: Una Stef, Getty Images

Source: "Bicyclus anynana 20110217 012300 (https://commons.wikimedia.org/wiki/File:Bicyclus_anynana_20110217_012300_5401M2.JPG) 5401M2 (https://commons.wikimedia.org/wiki/File:Bicyclus_anynana_20110217_012300_5401M2.JPG)"by Gilles San Martin is licensed under CC BY-SA 3.0 (https://creativecommons.org/licenses/by-sa/3.0/deed.en)

Figure 2. Wet (left) and dry (right) seasonal forms of *Bicyclus anynana*.

This butterfly has a phenotype that responds to the changing temperature conditions caused by global climate change. When it is cooler, the adults take on the form of 'dry-season' adults that live for a longer time and breed at the end of the season. Their wing patterns are similar to dry foliage so that they can escape predators. However, when the weather is warmer, the adults look more like 'wet-season' adults. These have a shorter life span but breed many times during this season. Their wing patterns have distinctive eye spots that prevent predation. As the seasons change, so does the butterfly's physical form.

Recessive disorders

Earlier we looked at the inheritance of the dominant trait of the PTC gene. Whether individuals can taste the bitter chemical or not does not compromise their health. However, there are other inherited dominant and recessive disorders that can have life-changing consequences.

Theory of Knowledge

How would you feel if you struggled to get health insurance or a job because of a known genetic disorder? The fear of discrimination is a concern for those that might want to get tested for genetic disorders. In many countries there are laws to protect such individuals. What is the situation in the country that you live in?

A recessive genetic condition means the individual would need to be homozygous recessive with two copies of the recessive allele to have the trait. One such recessive disorder is phenylketonuria or PKU. This affects approximately 1 in 24 000 live births globally and results in high morbidity. PKU is caused by a mutation in a gene on chromosome 12

Student view

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(**Figure 3**). This gene codes for an enzyme used in metabolism called phenylalanine hydroxylase (PAH) that converts the amino acid phenylalanine (Phe) into tyrosine (Tyr). The presence of one correct allele is sufficient to generate the functioning enzyme and results in a healthy individual.

In a child that is homozygous recessive for this allele, the Phe in the cell is not broken down and so toxic levels build up. The baby may initially seem healthy but within a few months, symptoms can develop, such as a musty odour from the skin and urine, fair skin, eczema, seizures, tremors and hyperactivity. In most countries around the world, babies are tested for PKU at 1–2 days of age. A simple heel stick gives a drop of blood that can be tested on a filter paper for the Phe:Tyr ratio. If the test reveals a high level of Phe, then further testing is conducted. If the condition is left untreated, brain damage can occur. Unfortunately, there is no quick treatment, but this condition can be well managed if a life-long dietary plan that keeps protein levels low is maintained, along with frequent blood tests.

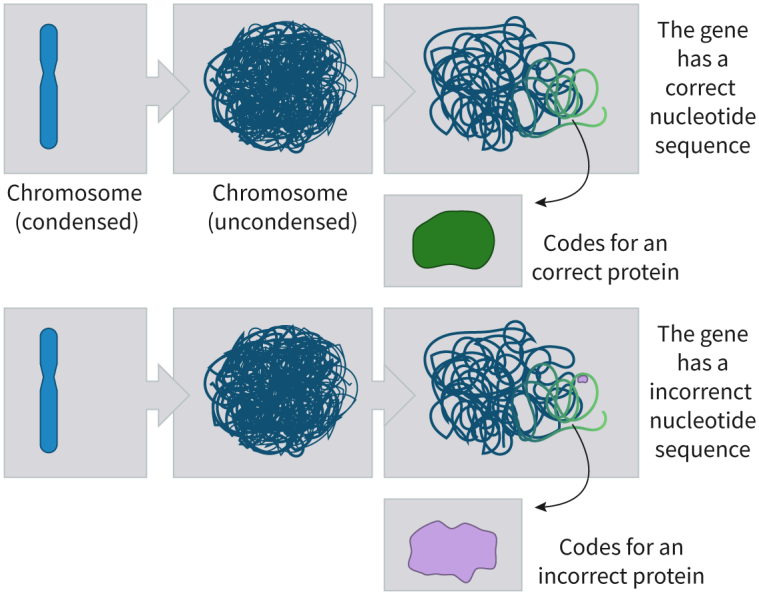

Figure 3. A mutation in the gene that makes the enzyme phenylalanine hydroxylase (PAH) causes a faulty enzyme to be generated. This can have serious consequences if left untreated.

More information for figure 3

The image is a diagram illustrating the relationship between chromosome structure, gene nucleotide sequence, and protein coding. It consists of two sections: the upper section representing a correct nucleotide sequence and the lower section representing an incorrect nucleotide sequence.

In the top section, a condensed chromosome depicted as a blue, rod-like shape is shown transitioning to an uncondensed chromosome, illustrated as a tangled blue line. This is followed by a more detailed depiction of the uncondensed chromosome with a clear nucleotide sequence. An arrow shows this correct sequence coding for a green, globular shape that represents a correct protein.

In the bottom section, a similar progression is illustrated. A condensed chromosome transitions to an uncondensed state with a different, improperly arranged nucleotide sequence. An arrow points from this sequence to a pink, irregular shape, indicating an incorrect protein.

Text annotations on the image provide these labels: "Chromosome (condensed), Chromosome (uncondensed)" on both sections, "The gene has a correct nucleotide sequence" and "Codes for a correct protein" on the top section, and "The gene has an incorrect nucleotide sequence" and "Codes for an incorrect protein" on the lower section.

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Student view

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Creativity, activity, service

Strand: Creativity and Service
Learning outcome: Demonstrate engagement with issues of global significance

Rare Disease Day

People with rare diseases such as PKU may struggle to get good health care and could be marginalised in a society.

With some members of your class, plan an event for Rare Disease Day. This falls on 29 February but you don't have to wait for a leap year! You can make an awareness campaign at any time.

You can use the information on this site (https://www.rarediseaseday.org/), or you can be creative.

Here are some ideas:

- You could make an infomercial to share at your school.
- You could make a fun quiz to share at your local supermarket.
- You could see if there are members of your community that might have a disorder and be willing to speak.
- If it is permitted in your country, you could raise funds to go towards a local charity or foundation.

In the activity below, you will work in groups to find out more about some recessive genetic conditions.

Activity

- **IB learner profile attribute:** Knowledgeable
- **Approaches to learning:**
  - Communication skills — Practising active listening skills
  - Social skills — Working collaboratively to achieve a common goal
- **Time required to complete activity:** 40 minutes
- **Activity type:** Group activity

Whole class *Jigsaw* activity with the class divided into groups of four. Each group will work together to research a recessive disorder. Examples could be PKU, sickle-cell disease, cystic fibrosis or Tay—Sachs disease. There should be the same number of disorders as there are groups.

Spend 15—20 minutes researching the disorder and take notes as you do this. After the 15—20 minutes, your teacher will give you a number: 1, 2, 3 or 4. Now you will join with people that have the same number as you (e.g. all the number 1s will join together, all the number 2s etc.). These new groups now should contain four students all knowledgeable about a different autosomal recessive disease. You now have a further 20 minutes for each student in the group to take turns to share their knowledge about their specific disorder. Use your active listening skills so that everyone has an opportunity to share the disorder they learned about. Take notes on the three that you did not study.

5 section questions

Allele combinations

Overview (/study/app/bio/sid-422-cid-755105/o

D3.2.8: Single-nucleotide polymorphisms D3.2.9: ABO blood groups D3.2.10: Incomplete dominance and codominance

Learning outcomes

By the end of this section you should be able to:

- Explain the ways that members of the same species can have variety in the gene pool.
- Describe the inheritance of different blood types.
- Compare and contrast the differences between incomplete dominance and codominance.

Look around at your classmates. How much of your DNA is the same as that of the person sitting next to you? You might be surprised to find out that you are 99.9% similar to this person! You might be at a school that has students from all parts of the globe and so look quite different. Even so, you are still genetically very similar! So what is it about the remaining 0.1% that gives such variety in the human gene pool? We will be studying what can be learned from these fascinating differences and how that helps us understand the patterns of disease in a population.

Single nucleotide polymorphism

Sometimes, a single nucleotide in the DNA gets randomly changed for another one. This is called a mutation. If this change is then passed on to the offspring, the new sequence can become prevalent in the population. Some of these differences are caused by a change of just one base in the DNA sequence. If at least 1% of the population has the same base letter at a certain position (called a variant), then that variant is known as a single nucleotide polymorphism or SNP (pronounced as *snip*). These SNPs are the most important and widespread kind of genetic variation found in biology. Estimates suggest that there are at least 80 million SNPs in the global human population. SNPs can be found throughout the genome. They can be in a gene that codes for a protein or in parts of the DNA that are non-coding and are responsible for other functions. The presence of SNPs can be examined and patterns have been found among different human populations. Pooling data of SNPs has meant that a database of SNP-associated traits can be built up to examine health, disease and drug response. For example, it is possible to examine a group of people that all have a particular disease and start to look at SNPs that are common to them all. Genome-wide association studies (GWAS) on specific diseases, using a large volume of metadata, can be investigated to start to understand their heritability.

Nature of Science

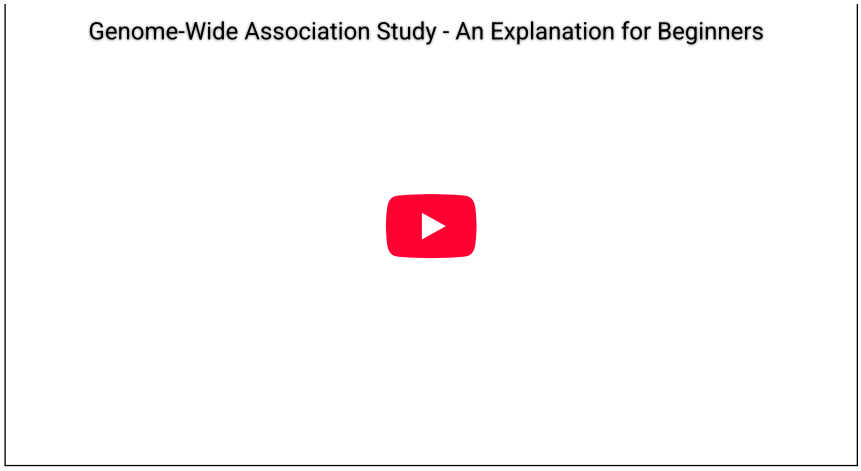
Aspect: Science as a shared endeavour

A large volume of metadata from thousands of sources has been used for GWAS and compiled into a freely available catalogue. This was started by the National Human Genome Research Institute (https://en.wikipedia.org/wiki/National_Human_Genome_Research_Institute) (NHGRI) in 2008. Since 2010 it has been a collaborative project with the European Bioinformatics Institute (https://www.ebi.ac.uk/gwas/diagram) (EBI). Thousands of pieces of data from publications are collated and sorted and can be viewed diagrammatically for their relationship to different forms of disease.

Watch **Video 1** to understand more about GWAS:

Student view

Overview (/study/ap 422-cid-755105/o



Video 1. An introduction to GWAS.

SNPs can then result in many different alleles for a single gene. The more SNPs within a gene, the more different alleles there will be. These multiple alleles available in a gene pool all add to the genetic variation possible. The different allelic forms of a gene are not necessarily permanent and can appear, and equally as easily disappear, in a population of organisms. To increase variation even further, each individual only inherits two alleles for a gene and so the combination of the alleles it possesses also relates to the variation.

Multiple alleles

Blood type is an example of a human trait controlled by multiple alleles and inherited from parents. It was only in 1900 when Landsteiner looked into why some blood transfusions were medically helpful while others gave an adverse, or sometimes even fatal, reaction that these different blood types were identified. The origin of these alleles is unknown but is thought to have evolved over several million years. The allele for blood type is found on chromosome 9 in the human genome and the gene is called the ABO gene. The three allelic forms A, B and O are each responsible for making their own type of enzyme (glycosyltransferase). This enzyme catalyses the production of specific carbohydrate chains (sugars) on the outside of the red blood cells. These sugars are capable of producing an immune response and so are referred to as antigens. So, it is the ABO type of gene that determines which kind of sugar (antigen) is made. However, each individual only has the possibility of two alleles for blood type.

The way that these blood types are written is as follows: type A (I^AI^A or I^Ai), type B (I^BI^B or I^Bi) or type O (ii). Someone that has one I^A allele and one i will have blood type A, as the I^A allele is dominant over i; they will have the A antigen on the surface of their red blood cells. An individual with one I^B allele and one i will have blood type B, as blood type B is dominant over O; they will have the B antigen on their red blood cells. However, a person that has two ii alleles (type O) will express neither antigen. Studies (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC9233352/) post COVID have shown that those with blood type A have a higher risk factor for contracting the disease while those with blood type O have a reduced risk.

There is one final alternative. Individuals that have alleles for glycosyltransferase A and glycosyltransferase B will express *both* antigen A and B on their red blood cells as both of these genes are equally dominant (codominant). This results in blood type AB (I^AI^B). **Table 1** summarises the four blood types.

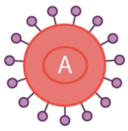
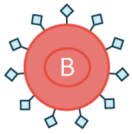
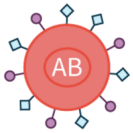
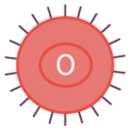
For each of these blood types with specific proteins on the outside of the red blood cells, there are specific antibodies in the plasma (**Table 1**). When giving blood transfusions, care must be taken, as it is the presence of antibodies in the plasma of the recipient that might result in clotting of the received blood. Clots are dangerous as they can break free and

Student view

Overview (/study/ap 422-cid-755105/o

move into other parts of the body; for example, they could block the blood flow to the brain or move into the lungs. So who can receive what kind of blood? A person with blood type A has type B antibodies in their own plasma. This means that if they received blood type B in a transfusion, their antibodies would clot the blood. Thankfully, such errors rarely happen in modern medical practice. The O blood type is known as the universal donor as this blood type has no antigens on the surface and so regardless of the blood type of the recipient, the antibodies would have nowhere to bind, and therefore would not cause clotting. A person with blood type AB is known as the universal recipient. Their red blood cells have both surface antigens, but these individuals have no antibodies in their plasma, and so can receive all blood types without hindrance.

Table 1. The four different blood types along with the corresponding antibodies present in the plasma.

	Type A	Type B	Type AB	Type O
Red blood cell type		 More information The image is a diagram featuring a large central circle labeled 'B'. Surrounding this central circle are multiple smaller nodes connected by lines. The nodes are placed symmetrically around the central circle, resembling a network or molecular structure. The diagram illustrates a central concept or node 'B' having associations or links to multiple surrounding elements. [Generated by AI]		

Student view

Overview (/study/ap 422-cid-755105/o

	Type A	Type B	Type AB	Type O
Antibodies in plasma	 Anti-B	 Anti-A	None	 Anti-A and Anti-B
Antigens in red blood cell	 A antigen	 B antigen	 A and B antigens	None

More information

The image depicts two symmetrical shapes representing Anti-A and Anti-B antibodies. Each shape consists of short lines arranged radially around a central point, forming a star-like design. The Anti-A antibody is shown in blue on the left, while the Anti-B antibody is in purple on the right. Below the illustrations, the text reads "Anti-A and Anti-B." The design suggests a visual comparison between the two types of antibodies based on their structural similarity.

[Generated by AI]

Codominance and incomplete dominance

As mentioned above, people that have the alleles for glycosyltransferase A and B will express both A and B antigens, resulting in blood type (I^AI^B). This is because these two alleles are codominant; neither is dominant over the other but both are expressed. This blood type is an example that you should be prepared to give as an example of codominance.

A different kind of dominance also exists in certain organisms and is known as incomplete dominance. This is when two parents with different phenotypes give rise to offspring with a phenotype that is intermediate between the two parental types. In the flower called the Marvel of Peru (*Mirabilis jalapa*), the allele for red flowers is not entirely dominant over the allele for white flowers. Therefore, in heterozygotes, a third phenotype – pink – results that is intermediate between the red and white flowers. **Figure 1** shows the three possible phenotypes for the Marvel of Peru.

The convention for incomplete dominance is to use a capital letter with a superscript letter for the allele. In this example, C^W is the white allele and C^R is the red allele. Using this notation, white flowers would have the genotype C^WC^W, red flowers would have the genotype C^RC^R and pink flowers would have the genotype C^WC^R. **Figure 2** shows how the intermediate flower colour is inherited in the Marvel of Peru.

Student view

Overview (/study/ap 422-cid-755105/o

Credit: Photos from Japan, Asia and othe of the world, Getty Images

Credit: Emon's photography orb, Getty Images

Credit: Nurma Agung Firmansyah, Getty Images

Figure 1. The three phenotypes of the Marvel of Peru flower: red, white and pink.

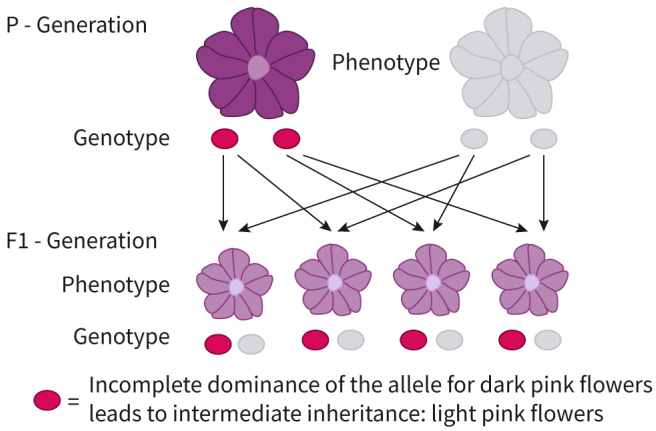


Figure 2. The inheritance of flower colour in the Marvel of Peru flower.

More information for figure 2

The diagram illustrates the inheritance pattern of flower color in the Marvel of Peru plant. At the top, there is the P-Generation showing two flower phenotypes: a pink flower on the left and a white flower on the right, with their respective genotypes represented by colored circles below them (pink and white circles respectively). Arrows from these genotypes point downwards to the F1-Generation.

Student view

Overview (/study/ap 422-cid-755105/o

In the F1-Generation, light pink flowers are shown in the middle as the resulting phenotype. The genotype is a combination of the parental genotypes, represented by one pink and one white circle. The diagram includes a legend at the bottom that explicates the concept of incomplete dominance, stating that the combination of alleles for dark pink flowers leads to intermediate inheritance, resulting in light pink flowers.

[Generated by AI]

Try playing a blood typing simulation in the activity below to help with your understanding of blood groups.

Activity

Section Student... (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/from-haploid-to-diploid-id-45747/print/) Assign

- **IB learner profile attribute:** Inquirer
- **Approaches to learning:** Thinking skills — Asking questions and framing hypotheses based upon sensible scientific rationale
- **Time required to complete activity:** 20 minutes
- **Activity type:** Individual activity

Section Student... (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/expression-of-phenotypes-id-46204/print/) Assign

Try this Blood Typing (https://www.accessdl.state.al.us/AventaCourses/access_courses/virtuallabs_ua_v21/01_unit/01-01/01-01_learn.htm) lab simulation. As a lab technician you can help a patient needing a blood transfusion by selecting the most appropriate donor.

5 section questions

D3. Continuity and change: Organisms / D3.2 Inheritance

Genetics of the Y chromosome

D3.2.11: Sex determination in humans D3.2.12: Haemophilia D3.2.13: Pedigree charts

Learning outcomes

By the end of this section you should be able to:

- Describe that the sperm determines sex in humans.
- Describe haemophilia as an example of a sex-linked genetic disorder.
- Illustrate how pedigree charts are used to determine inheritance in family members.

Determining sex

Expectant parents are often very excited to know the gender of their unborn child and will go to great lengths to try to determine it. Almost all cultures have stories of how to predict what is being carried by the mother. An ancient Egyptian text (https://www.smithsonianmag.com/smart-news/egyptian-papyrus-reveals-old-wives-tale-very-very-old-indeed-180970066/#:~:text=To%20find%20out%2C%20the%20woman,which%20grain%20signifies%20which%20sex.) tells of

Student view

Overview (/study/ap 422- cid- 755105/o

pregnant women urinating on a bag of barley and then on a bag of wheat. If the barley sprouted first it was a boy and if the wheat sprouted first it was a girl! So, what is the genetic reason for whether a baby boy or girl is formed at the time of fertilisation? We will find out in this section.

We now know that there is an equal chance of conceiving a boy or a girl at every pregnancy. Sex determination is inherited. In humans, the 23rd pair of chromosomes are the sex chromosomes. A haploid egg contains one copy of all 23 chromosomes. The 23rd chromosome will be an X chromosome. A haploid sperm also has one copy of all 23 chromosomes but in this case the 23rd chromosome will be either an X or a Y chromosome. Either could fertilise the egg, and therefore it is the sperm that ultimately determines sex. When fertilisation occurs, the zygote will contain the chromosomes that were carried within the gametes. In humans, two X chromosomes will result in a female and an X and Y chromosome will result in a male. Other species have different ways of determining gender. In the Komodo dragon (*Varanus komodoensis*), for instance, the similar chromosomes are found in the male as ZZ. The female has the dissimilar chromosomes of ZW.

Sex determination in humans is illustrated in **Figure 1**.

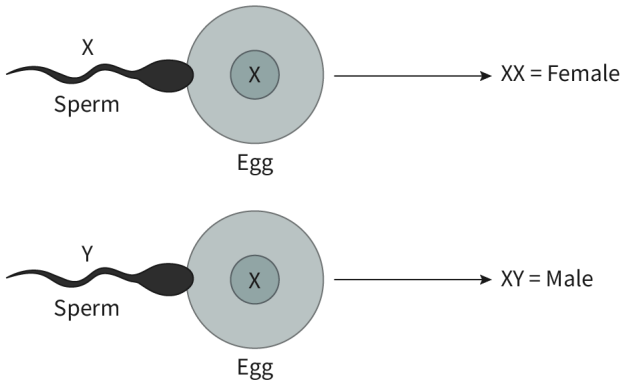


Figure 1. The type of sperm (X or Y) that fertilises the egg determines the gender of the zygote.

More information for figure 1

This diagram illustrates the role of sperm in determining the gender of a human zygote. There are two main parts of the diagram, each showing a different type of sperm interacting with an egg.

In the top section, an X chromosome-bearing sperm is depicted approaching and fertilizing an egg that already contains an X chromosome, resulting in an XX chromosome combination. Arrows indicate the development of a female zygote.

In the bottom section, a Y chromosome-bearing sperm is fertilizing an egg, also with an X chromosome, leading to an XY combination. Arrows show the development of a male zygote.

The diagram points out the significance of the sperm type (X or Y) in determining whether the resulting zygote will be male or female. Text annotations clarify the sperm types and the outcome of each fertilization scenario (XX = Female, XY = Male).

[Generated by AI]

Student view

A particular gene on the Y chromosome called the SRY gene is important in allowing the zygote to develop into a male embryo. This gene encodes for a sex-determining region Y protein which acts as a transcription factor and binds to

Overview (/study/ap 422-cid-755105/o

particular parts of the DNA to control the expression of certain genes. This factor enables a foetus to develop male gonads and prevents the development of female organs.

Sometimes the meiotic division responsible for making eggs or sperm at oogenesis or spermatogenesis goes wrong. This is called non-disjunction. If this happens in the 23rd pair, an incorrect number of sex chromosomes can result. Klinefelter syndrome is one of the most common example of aneuploidy (**Figure 2**). Aneuploidy is when an individual has an extra chromosome(s) or is missing a chromosome. This is different from polyploidy where there are extra complete sets of chromosomes in the nucleus. Klinefelter syndrome occurs in about 85–250 cases for every 100 000 males born and can result in a person having 47 (XXY), 48 (XXXY) or 49 (XXXXY) chromosomes.

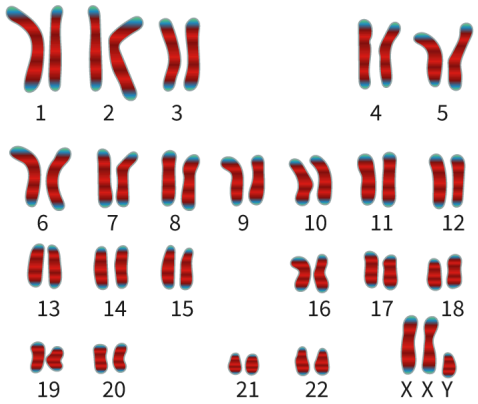


Figure 2. A karyogram of a male with Klinefelter syndrome. This person has 47 chromosomes (XXY).

More information for figure 2

The image displays a karyogram of human chromosomes arranged in pairs from 1 to 22, followed by the sex chromosomes labeled as X and Y. Each chromosome pair is visually represented with two chromatids, except for the 23rd pair, which shows an additional X chromosome, indicating an XXY configuration typical of Klinefelter syndrome. The chromosomes are color-coded with red and blue shading, enhancing their banding patterns, which are used to differentiate and identify individual chromosomes. The structure is orderly, allowing for analysis of size, banding, and presence of extra or missing chromosomes.

[Generated by AI]

Sex-linked genetic disorders

The X chromosome is about three times larger (https://www.genome.gov/about-genomics/fact-sheets/X-Chromosome-facts) than the Y chromosome and has about 900 genes compared with the 55 genes found on the Y chromosome. With a greater number of genes comes a greater possibility of mutations, and therefore disorders. Due to the difference in sizes, there are portions of the X chromosome that have no homologous Y counterpart (**Figure 3**). This means that if an allele is present for a disorder on the X chromosome only, the individual would then be positive for that disorder.

Student view

Overview (/study/ap 422-cid-755105/o

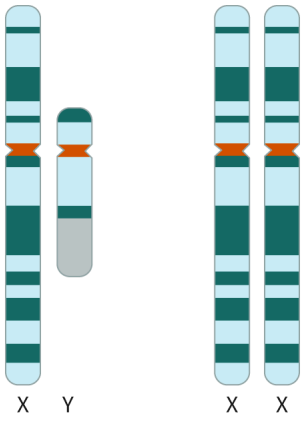


Figure 3. In a human male, the X and Y chromosomes are a different size. Genes that are present on the X may not be present on the Y.

More information for figure 3

The image shows an illustration comparing the structure of human sex chromosomes in both males and females. The left side depicts a male with one X and one Y chromosome. The Y chromosome is significantly shorter and has fewer dark bands compared to the X chromosome, illustrating the difference in size and gene density. The right side shows a female with two X chromosomes, which are alike in size and banding pattern, indicating they are homologous pairs. This visual representation highlights that the X chromosome is larger, with more genetic information, compared to the Y chromosome.

[Generated by AI]

Certain alleles that confer inherited conditions are found on the long arm of the X chromosome. These are called sex-linked genetic disorders and include colour blindness and haemophilia.

In haemophilia, blood-clotting factors known as Factor XIII or Factor IX are made in insufficient quantities. This is often caused by a mutation in a gene for one of these clotting factors. This means that when an individual is cut, they can bleed excessively, even after a relatively small trauma. Bleeding might not be visible and can be internal, which is when damage occurs to the blood vessels inside the body; this can be equally life threatening. It has been called the 'disease of the kings' as Queen Victoria of England was a carrier, however no living member of the current royal family carries the defective gene. New gene therapy technology has been used with success for haemophilia. The gene that makes Factor IX has been inserted into a virus that infects humans but does not cause disease. This virus is called a vector and can be used to infect liver cells that are responsible for making the clotting factor. Functional protein is then generated by these cells and so this removes the need for injections of Factor IX every three to four days for life.

Study skills

Notation in science is important for clarity. These sex-linked disorders are written out using superscript letters.

It is convention to use the letter 'H' when talking about haemophilia.

X^HX^H = unaffected female

X^HX^h = carrier female for haemophilia

X^hX^h = affected female for haemophilia

X^HY = unaffected male

X^hY = affected male for haemophilia

Student view

Overview (/study/ap 422-cid-755105/o

Pedigree charts and the pattern of inheritance

In science, models are often used to help explain complex concepts. Pedigree charts can be used to illustrate the inheritance of a trait through a family's history. These are useful when there is some knowledge about family members that have been positive for a disease and they can be placed onto the chart. Such a chart can then help make predictions about the likelihood of future generations inheriting a disorder.

Pedigree charts use common features but should have a key to clarify their meaning. Circles generally refer to females and squares are used for males. White circles or squares mean that the person is unaffected for a trait and a black circle or square means that they are affected. Some individuals that are unaffected may still carry the recessive allele, and can therefore pass it on to their offspring. We call these individuals carriers. Using the information about the affected individuals in a chart, deductive reasoning is needed to discern the genotypes of other members in the pedigree.

Nature of Science

Aspect: Observations

Pedigree charts are used by scientists to show a pattern of inheritance clearly. Examination of several generations enables the genotypes of specific individuals to be deduced. This kind of deductive reasoning allows scientists to draw conclusions from a theory they have formed from observations.

There are specific patterns that can be observed in pedigree charts and these should be looked for. In charts showing autosomal recessive traits, the males and females might be equally represented for having the trait (**Figure 4**). Such a trait can only be expressed if the individual is homozygous recessive. If both parents are affected, the offspring should be also. If the parents are both unaffected, but the offspring has the trait, then the parents must be heterozygous. The trait can skip generations.

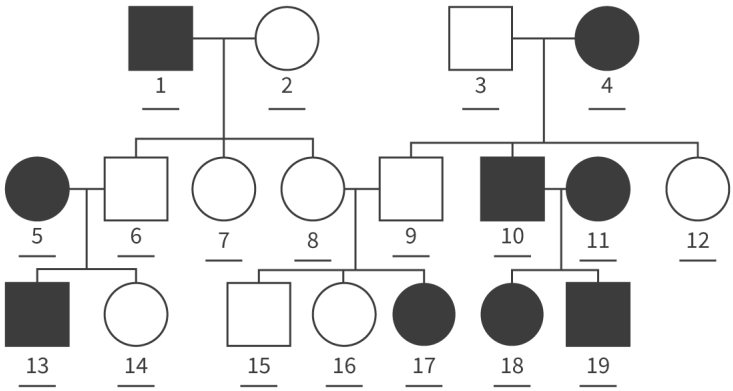


Figure 4. Pedigree chart showing autosomal recessive inheritance.

More information for figure 4

This pedigree chart represents autosomal recessive inheritance within a family. The chart consists of shapes representing family members: squares for males and circles for females. Shaded shapes indicate individuals expressing the trait, while unshaded shapes represent those not expressing the trait. The chart visually depicts two generations with connected lines showing parent-child relationships. Many members in the first generation are unaffected, while some in the second generation are affected, demonstrating that the trait can skip generations and is expressed only when individuals are homozygous recessive.

Student view

[Generated by AI]

Overview (/study/ap 422-cid-755105/o

In charts showing autosomal dominant traits, the trait does not skip generations (**Figure 5**). If a child has the trait, at least one parent must also have the trait. If neither of the parents have the trait, the offspring will not exhibit it either. If both parents do have the trait, but the offspring does not, then the child will be homozygous recessive and the parents will be heterozygous or carriers.

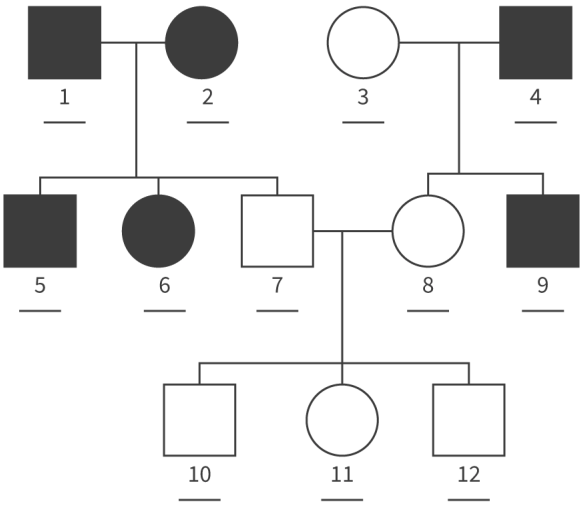


Figure 5. Pedigree chart showing autosomal dominant inheritance.

More information for figure 5

The image is a pedigree chart illustrating autosomal dominant inheritance. Symbols in the chart represent family members across three generations, with circles indicating females and squares indicating males. Shaded symbols represent individuals exhibiting a specific genetic trait, indicative of a dominant inheritance pattern. In this chart, the trait appears in each generation without skipping, consistent with autosomal dominance.

Starting at the top, individual 1 is a male and individual 2 is a female, both exhibiting the trait. They have a child, individual 5 (male), who also exhibits the trait. Next to them are individuals 3 and 4, who do not have the trait.

Moving down, individuals 6 and 7 form a pair, where individual 6 (female) exhibits the trait while individual 7 (male) does not. Their offspring, individuals 10 through 12, show a mix of trait expression. Individual 10 (male) and 12 (male) do not exhibit the trait, while individual 11 (female) does.

Additionally, individuals 8 and 9 form another pair, with individual 9 (male) exhibiting the trait, while individual 8 (female) does not. This chart effectively illustrates how a genetic trait can be inherited in an autosomal dominant pattern across generations, where at least one parent must pass on the trait for it to be expressed in offspring.

[Generated by AI]

Section Student (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/allele-combinations-id-46205/print/) Assign

Maintaining high genetic variation in a population is important as it results in a species being able to adapt to environmental changes. Breeding within a small population reduces a species' variation. Any genetic diseases within a smaller gene pool will then be more likely to be observed. Throughout history, many human cultures have favoured consanguineous marriage (marriage between two blood relations that are second cousins or closer) as this is seen to help retain a strong lineage. This cultural practice can lead to the increased presence of autosomal recessive disorders, as well as congenital abnormalities. Many societies now have restrictions over close family marriage for this reason.

Student view

Overview (/study/ap 422-cid-755105/o

Try the activity below to analyse pedigree charts in an online worksheet.

Activity

- **IB learner profile attribute:** Caring
- **Approaches to learning:** Thinking skills — Applying key ideas and facts in new contexts
- **Time required to complete activity:** 45 minutes
- **Activity type:** Individual activity

Use this website from HHMI (https://www.biointeractive.org/classroom-resources/analyzing-pedigrees) and click on Analysis document pdf (top right) to go to the worksheet on Pedigrees. Work through the sheet with a partner and become familiar with viewing many different pedigree charts in new disorders and contexts. Use this knowledge to become more compassionate to those that are suffering with certain genetic disorders.

5 section questions

D3. Continuity and change: Organisms / D3.2 Inheritance

Variation in populations

D3.2.14: Continuous variation D3.2.15: Box-and-whisker plots

Learning outcomes

By the end of this section you should be able to:

- Distinguish between continuous variation such as skin colour and discrete variation such as ABO blood group.
- Illustrate continuous variables using box-and-whisker plots.

Discrete and continuous variation

The inheritance of traits such as skin colour is neither good nor bad and has no moral connection. The inheritance of skin colour is purely the result of natural selection. What is the purpose of the global ‘sepia rainbow’ of the human population and how are the different colours generated? This will be a focus in this section.

So far, we have discussed genetic traits that are controlled by one gene. The presence or absence of these alleles means that organisms in a population can be put easily into just a few distinct groups. This discrete variation refers to traits where individuals are clearly one phenotype or another. Blood types are discrete, as an individual is blood type A, B, O or AB (**Figure 1**). There are no gradual changes between these types and no in-betweens.

Student view

Overview (/study/ap 422-cid-755105/o

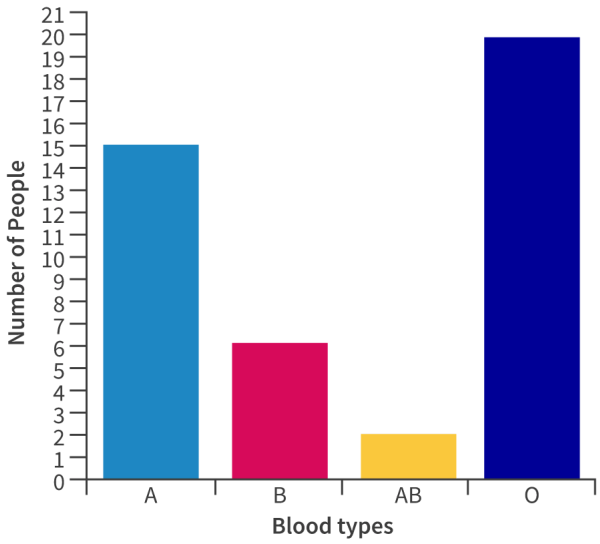


Figure 1. A bar graph showing discrete variation of blood types.

More information for figure 1

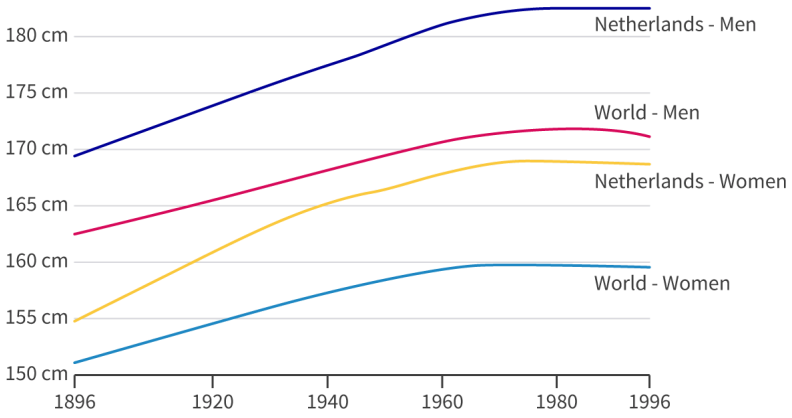
The bar graph illustrates the distribution of blood types in terms of the number of people. The X-axis represents discrete blood types labeled as A, B, AB, and O. The Y-axis indicates the number of people, ranging from 0 to 21.

- Blood type A is represented by a bar reaching up to 15 people.
- Blood type B has a bar that reaches up to 5 people.
- Blood type AB reaches 2 people.
- Blood type O reaches the highest at 20 people.

This graph shows that blood type O is the most common, followed by A, with AB being the least common.

[Generated by AI]

However, there are other traits, like human height for example, where there are no specific groups, but show a gradual change between two extremes in a population. For many years, the Dutch have been shown to be the tallest nation on Earth, although the reasons are not understood. If we compare people from two different generations, we see that in 1980 men and women were both 5.3 cm taller than the men and women born in 1930. These ideas are illustrated in **Figure 2**.



Student view

Overview (/study/ap 422-cid-755105/o

Figure 2. Men and women in the Netherlands are considerably taller than the global average.

More information for figure 2

The graph represents the trends in average heights for men and women in the Netherlands compared to the global average from 1896 to 1996. The X-axis represents the years spanning from 1896 to 1996, while the Y-axis represents height in centimeters, ranging from 150 cm to 180 cm.

Four lines are depicted: 1. **Netherlands - Men**: This curve rises from around 165 cm in 1896 to approximately 183 cm by 1996, showing a consistent increase in height. 2. **World - Men**: This line moves from about 162 cm to around 173 cm over the same period, also indicating a rising trend but at a slower rate compared to Dutch men. 3. **Netherlands - Women**: Beginning at approximately 155 cm in 1896, this line rises to about 170 cm by 1996, showing a steady increase. 4. **World - Women**: This trend starts at roughly 152 cm and grows to around 160 cm, showing a slower rate of increase than Dutch women.

The overall pattern shows that Dutch men and women are taller than the global average women and men throughout the period, with a notably larger increase in height over time.

[Generated by AI]

Polygenic inheritance

Height is not controlled by one gene alone; in fact, it is controlled by many genes interacting together. This is called polygenic inheritance. Environmental factors such as diet can also contribute to how tall a person will be at adulthood. Imagine surveying all the people in your town that are the same age and recording the height of each to the nearest cm. You could then make a frequency chart of the different height brackets and produce a frequency histogram. You could also calculate the mean, median and mode of this population. You would find that there were a small number of people at either extreme, being either very short or very tall. However, most people would fall around the mean. This continuous variation results in a spread called a normal distribution and, when plotted, results in a bell-shaped curve. All continuous variation in a population can be seen to have a normal distribution.

Another example of continuous variation is human skin colour. This is also controlled by many different genes (**Figure 3**).

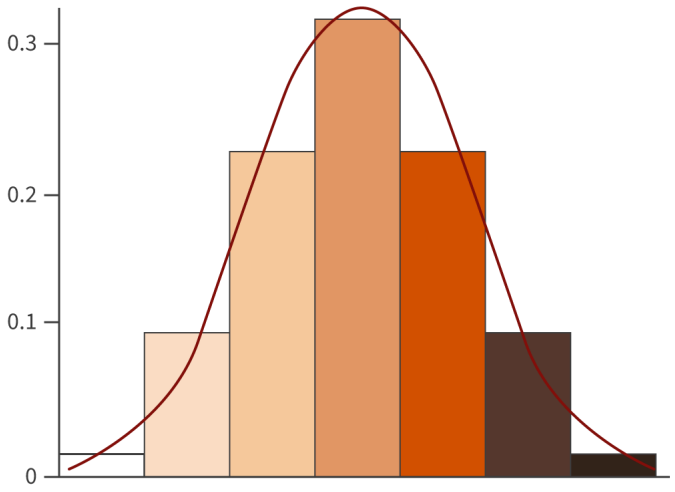


Figure 3. A bar graph showing continuous variation of skin colour.

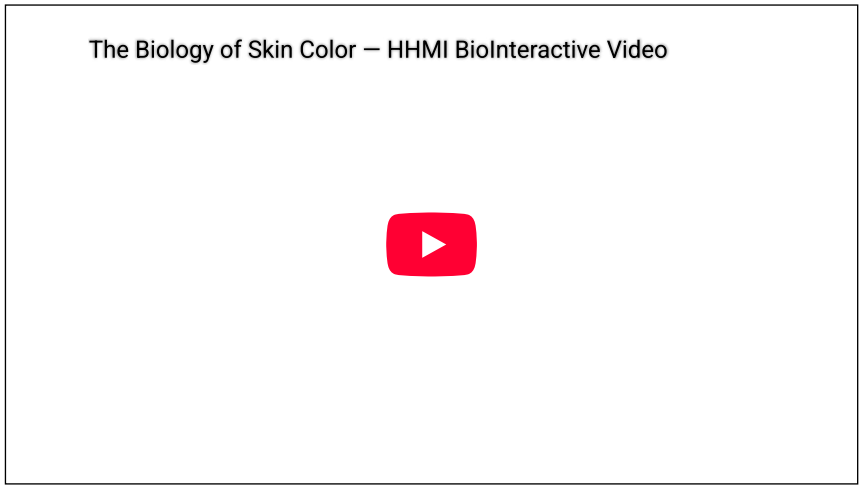
Student view

More information for figure 3

This bar graph demonstrates the continuous variation in human skin color. The X-axis represents different skin color categories, ranging from light to dark hues. The Y-axis shows the frequency or proportion of individuals, ranging from 0 to 0.35. There are several bars, each with different heights indicating the distribution of skin colors in a population. A curve overlays the bars, depicting a normal distribution, with the highest frequency bar in the center, representing the most common skin color range. The trend indicates a gradual increase in frequency towards the median skin colors, followed by a decrease towards the less common extreme ends of the spectrum.

[Generated by AI]

The vast range of skin colours in the human population is due to the presence or absence of melanin. Some genes encode for melanin and some do not. It is the additive effects and combination of all these alleles in an individual that means they have the skin tone that they do. Even siblings with the same parents can have quite different skin colours purely because the combination of alleles they inherited from their parents is unique to them. Examination of people that are originally from various parts of the world helps to explain why skin colour has evolved. The more genes that express melanin, the darker the skin colour. This melanin protects the nucleus from dangerous UV wavelength. Anthropologist Nina Jablonski correlated the amount of UV present at a location on the planet with the skin colour of the people who live there. Where UV is strong, the skin is dark. Where the UV is less strong, skin is lighter. Jablonski used the phrase 'this beautiful sepia rainbow' to describe the many possible shades of skin colour. Watch **Video 1** to learn more about this.



Video 1. The 'beautiful sepia rainbow' of skin colour.

Box-and-whisker plots

Visualising data clearly is really important for scientists to be able to analyse it properly. You may have two data sets that have the same mean, but the distribution of those data could be quite different. We can use box-and-whisker plots to effectively graph the variation of a trait. In traits that are continuous and show a large range, the box plot allows this to be seen clearly.

Let's examine a data set such as these numbers:

14, 15, 4, 6, 4, 10, 12, 7, 11

First put the data in ascending order from smallest to largest:

4, 4, 6, 7, 10, 11, 12, 14, 15

As this data set has an odd number of values (nine), to calculate the median, you take the middle (or fifth) number which is 10. If the data set has an even number of values, then you add up the two middle numbers and divide by two.

Next identify the first quartile (Q1) by looking at the four numbers below the median (4, 4, 6, 7). As it is an even number of values, take the sum of the middle two values (4 + 6 = 10) and divide by 2 = 5.

Then calculate the third quartile (Q3) by looking at the four highest numbers (above the median: 11, 12, 14, 15). As it is an even number of values, Q3 is found by taking the sum of the middle two values (12 + 14 = 26) and dividing by 2 = 13.

Next calculate the interquartile range (IQR) by subtracting Q1 from Q3:

$$\begin{aligned} \text{IQR} &= \text{Q3} - \text{Q1} \\ &= 13 - 5 \\ &= 8 \end{aligned}$$

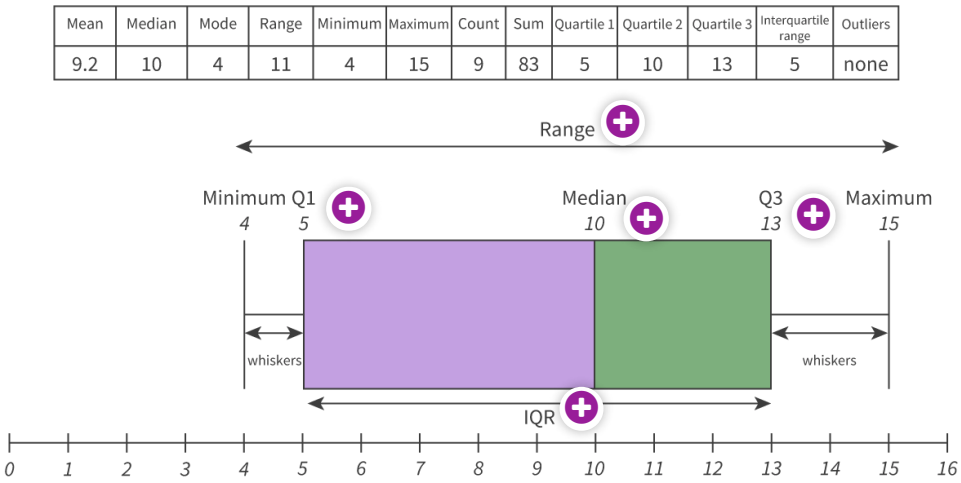
The information in **Table 1** can all be calculated for our data set.

Table 1. Values calculated for the box-and-whisker plot from the data set given.

Statistical Parameter	Value
Mean	9.2
Median	10
Mode	4
Range	11
Minimum	4
Maximum	15
Count	9
Sum	83
Quartile 1	5
Quartile 2	10
Quartile 3	13
Interquartile range	8
Outliers	none

In the box-and-whisker plot shown in **Interactive 1**, you can see that the line that is drawn within the box is the median: 50% of the data is less than this and 50% is above this. It also shows the second to third quartile as the ‘box’ with the minimum and maximum as ‘whiskers’ extending from the box.

Overview (/study/ap 422-cid-755105/o



Interactive 1. Generation of a Box-and-whisker Plot from the Data Set Given.

More information for interactive 1

The interactive is a combination of a box-and-whisker plot and a summary table of descriptive statistics. It also contains hotspots that provide additional information.

The table of descriptive statistics is provided at the top. At the bottom is a number line from 1 to 16. A box is drawn from the value 5 on the number line to 13.

The summary table provides key statistics for the dataset and shows the following values: Mean: 9.2, Median: 10, Mode (most frequent value): 4, Range $(\text{Max} - \text{min} = 15 - 4)$: 11, Minimum: 4, Maximum: 15, Count (Number of data points):9, Sum of all values: 83, Quartile 1 (Q1): 5, Quartile 2 (Q2): 10, Quartile 3 (Q3): 13, Interquartile range (IQR): 8 $(13 - 5)$, and outliers as none.

The box-and-whisker plot shows the following:

1. Minimum (Lower whisker):4, First Quartile (Q1, left edge of the box):5. The hotspot of Q1 reads “To calculate Q1, we take the four numbers below the median (4, 4, 6, 7). As it is an even number of values, take the sum of the middle two values $(4 + 6 = 10)$ and divide by $2 = 5$.”

2. Median (the vertical line inside the box):10, the hotspot of the median reads “As this data set has an odd number of values, to calculate the median, you take the middle number which is 10.”

3. Third Quartile (Q3, right edge of the box):13, the hotspot of Q3 reads “To calculate the Q3, we take the four highest numbers above the median (11, 12, 14, 15). As it is an even number of values, Q3 is Q2 found by taking the sum of the middle two values $(12 + 14 = 26)$ and dividing by $2 = 13$.”

4. Interquartile range (IQR, width of the box):8, the hotspot reads “To calculate the interquartile range (IQR) subtract Q1 from Q3.

$IQR = Q3–Q1 = 13–5 = 8$.”

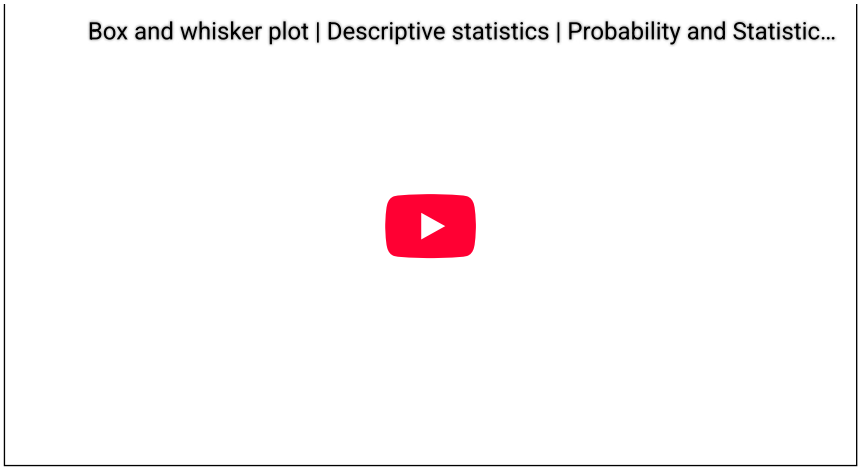
5. The whiskers extend between the values 4 and 5 and also between 13 and 15.

6. “Range” has a hotspot that reads “Calculate the range by subtracting the smallest value (4) from the largest value (15).”

We observe that the median (10) is higher than the mean (9.2) indicating a left-skewed distribution, the mode is 4, which is the minimum value, indicating clustering at the lower end. The IQR (88) shows a moderate spread in the middle 50% of the data.

Student view

Overview (/study/ap 422-cid-755105/o



Video 2. Generation of a box-and-whisker plot.

Creativity, activity, service

Strand: Creativity, Service
Learning outcome: Demonstrate the skills and recognise the benefits of working collaboratively

Use the concept of continuous variation of skin colour as an impetus to celebrate diversity at your school.

Use information from the United Nations (particularly the UN's Sustainable Development Goal #10) to help you collaborate and plan an assembly for End Racism Day (https://www.un.org/en/observances/end-racism-day) on 21 March.

If your school is not very diverse, you could make an infomercial to educate others about the dangers of racism.

Try the activity below to generate and analyse data on the continuous variable of height.

Activity

- **IB learner profile attribute:** Knowledgeable
- **Approaches to learning:** Communication skills — Presenting data appropriately
- **Time required to complete activity:** 30 minutes
- **Activity type:** Group activity

You are going to record variation in handspan to the nearest centimetre.

The handspan is the measurement from the tip of the thumb to the tip of the little finger when your hand is spread open. You can measure this by spreading your hand as wide as possible on a ruler.

You should record all the data in a document shared with the whole class.

In pairs, enter the data into a box-and-whisker generator like this (https://www.statskingdom.com/advanced-boxplot-maker.html).

Compare your plots with a neighbouring pair to ensure that you have calculated this correctly. Be able to explain what the box-and-whisker plot shows.

Student view

Unlinked genes (HL)

Higher level (HL)

Learning outcomes

By the end of this section you should be able to:

- Explain how unlinked genes segregate and assort independently in meiosis.
- Predict the inheritance of pairs of unlinked genes in dihybrid crosses.
- Predict genotypic and phenotypic ratios in dihybrid crosses of unlinked autosomal genes using Punnett grids.

Unlinked dihybrid cross

How can two grey-haired, black-eyed rabbits produce white-haired, red-eyed offspring? When genes are found on the same chromosome, and are close together, they are said to be linked. If they are on different chromosomes, they are unlinked. **Figure 1** illustrates linked and unlinked genes.

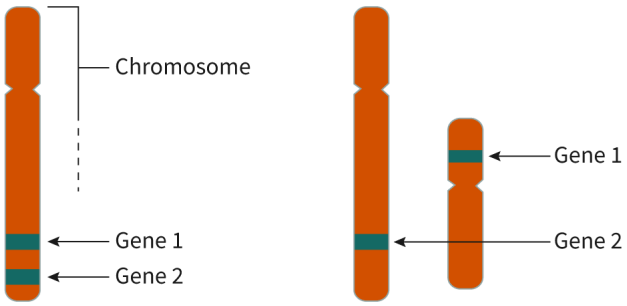


Figure 1. Linked genes can be found on the same chromosome while unlinked genes are on different chromosomes.

More information for figure 1

The image is a diagram illustrating linked and unlinked genes on chromosomes. On the left, there is a single chromosome with two genes labeled "Gene 1" and "Gene 2" positioned closely together, representing linked genes. On the right, there are two separate chromosomes, each featuring one gene: "Gene 1" on one chromosome and "Gene 2" on the other, illustrating unlinked genes. The diagram visually represents the concept of linkage and its impact on genetic inheritance.

[Generated by AI]

Student view

The fact of whether genes are linked or unlinked becomes important when we start to think about how genes end up in gametes in the process of meiosis. In this section we will focus on unlinked genes. When the gametes

are being made in meiosis, pairs of alleles for these different genes will separate (segregate) and then join together (assortment) independently from one another. We can say that the inheritance of one allele is independent of the inheritance of the other and so is known as independent assortment. For ease, we can look at what happens in meiosis in a diploid cell with just two pairs of chromosomes (https://www.sumanasinc.com/webcontent/animations/content/independentassortment.html).

The resulting gametes have all possible combinations of the alleles. In the linked example, only two pairs of chromosomes are shown. In the human genome, there are 23 pairs of chromosomes and so even in one individual, there are a vast number of possible combinations of alleles! This is why meiosis through sexual reproduction leads to increased genetic variation.

When we considered Mendel's breeding experiments with pea plants in an earlier section, we were looking at the inheritance of *single* traits in a monohybrid cross. Now let's consider the inheritance of *two* unlinked autosomal genes for two separate traits. We can use a dihybrid cross for the inheritance of two traits and so predict what the offspring might inherit from their parents.

Let's imagine we have some rabbits and we are to examine two traits – coat colour and eye colour – that we understand are unlinked (on separate chromosomes). Some of the rabbits have a grey coat and some have a white coat; some have red eyes and some black eyes.

We are told that grey is dominant over white hair and that black eyes are dominant over red. We can use the following notation:

G = grey, g = white

B = black, b = red

We can decide to do a breeding experiment. In the parent (P) generation we use pure breeds (where the parents are homozygous for both traits).

The male is a pure-bred grey-haired, black-eyed rabbit = GGBB

The female is a pure-bred white-haired, red-eyed rabbit = ggbb

Firstly, we need to consider what kind of gametes each rabbit can make. Think back to the animation earlier and the gametes that are made by segregation and independent assortment. In this case, the gametes made are limited. In meiosis, our male can only make four identical gametes containing the dominant alleles GB.

The female, by meiosis, can only make four identical gametes all with the recessive alleles gb.

We can show this in a 4 × 4 Punnett grid. Convention says that we put the same letters together, with the capital letter denoting the dominant allele first. All the offspring of the F1 generation would have a genotype of GgBb; they are heterozygous for both traits. The phenotype of all these rabbits is the same: they are all grey-haired and black-eyed. **Figure 2** illustrates this cross.

Overview (/study/ap 422- cid- 755105/o

	Male rabbit			
Female rabbit	GB	GB	GB	GB
gb	GgBb	GgBb	GgBb	GgBb
gb	GgBb	GgBb	GgBb	GgBb
gb	GgBb	GgBb	GgBb	GgBb
gb	GgBb	GgBb	GgBb	GgBb

Figure 2. Dihybrid cross of pure-bred grey-haired, black-eyed rabbit with a pure-bred white-haired, red-eyed rabbit.

More information for figure 2

The image is a Punnett square used to illustrate a dihybrid cross between two rabbits. The grid is labeled with genetic information. The top row represents a male rabbit with the alleles 'GB', and the left column represents a female rabbit with the alleles 'gb'. The cells within the grid demonstrate the resulting genotypes of the F1 generation, all of which are 'GgBb', indicating that the offspring are heterozygous for both traits. An illustration of a rabbit is shown in the top corner of the grid around the labels for the male and female rabbits, emphasizing the subject of the cross. Each cell in the 4x4 grid shows the same genotype 'GgBb', which indicates all offspring will have a phenotype expressing dominant traits, specifically grey hair and black eyes.

[Generated by AI]

Now let's try a different cross. This time we will breed a male and female from the F1 generation.

A male rabbit has the genotype GgBb and so first we have to consider what gametes he can make in meiosis. Remember that each allele could segregate and recombine independently of any other allele, so the gametes made could be GB, Gb, gB or gb.

A female rabbit from the above cross also has the genotype GgBb and so could also make the gametes GB, Gb, gB or gb.

This cross results in a 9:3:3:1 ratio of the different phenotypes. This second cross is illustrated in **Figure 3**.

	Male rabbit			
Female rabbit	GB	Gb	gB	gb
GB	GGBB	GGBb	GgBB	GgBb
Gb	GGBb	GGbb	GgBb	Ggbb
gB	GgBB	GgBb	ggBB	ggBb
gb	GgBb	Ggbb	ggBb	ggbb

	Grey haired; black eyed	9
	Grey haired; red eyed	3
	White haired; black eyed	3
	White haired; red eyed	1

Figure 3. Two heterozygote rabbits from the F1 generation are bred together to give a 9:3:3:1 ratio of phenotypes.

More information for figure 3

Student view

Overview (/study/ap 422-cid-755105/o

The image is a Punnett square diagram illustrating the results of breeding two heterozygote rabbits from the F1 generation. The diagram is a square grid with columns representing the gametes from the male rabbit and rows representing gametes from the female rabbit. Each cell within the grid shows a possible genotype outcome of the breeding, based on the genetic contributions from each parent.

Along the top, the male rabbit's gametes are labeled as GB, Gb, gB, and gb, while the female rabbit's gametes are labeled similarly down the left side. The resulting genotypes in the grid are marked for each combination, such as GGBB, GGBb, GgBB, etc. The background colors of these cells correspond to specific phenotypic outcomes labeled below the grid:

- Blue indicates grey-haired, black-eyed rabbits with a count of 9
- Green indicates grey-haired, red-eyed rabbits with a count of 3
- Purple indicates white-haired, black-eyed rabbits with a count of 3
- Red indicates white-haired, red-eyed rabbits with a count of 1

[Generated by AI]

A final breeding experiment can be done by crossing a male rabbit heterozygous for both traits with a female rabbit homozygous for both traits, as shown in **Figure 4**. This cross leads to a phenotypic ratio of 1:1:1:1.

	Male rabbit			
Female rabbit	GB	Gb	gB	gb
gb	GgBb	Ggbb	ggBb	ggbb
gb	GgBb	Ggbb	ggBb	ggbb
gb	GgBb	Ggbb	ggBb	ggbb
gb	GgBb	Ggbb	ggBb	ggbb

	Grey haired; black eyed	4
	Grey haired; red eyed	4
	White haired; black eyed	4
	White haired; red eyed	4

More information

The image is a Punnett square diagram used to illustrate a rabbit breeding experiment, showcasing the genetic outcomes when crossing a male rabbit heterozygous for both traits with a female rabbit homozygous for both traits. The diagram aligns the genotypes of each parent along the top (male: GB, Gb, gB, gb) and side (female: gb) to calculate the potential genotypes of the offspring in the grid cells.

Key: - GgBb (light blue): Grey haired; black eyed - Ggbb (lime green): Grey haired; red eyed - ggBb (purple): White haired; black eyed - ggbb (pink): White haired; red eyed

Each color-coded genotype corresponds to a phenotypic trait combination and frequency of 4, illustrating a classic 1:1:1:1 phenotypic ratio. This visual representation supports the concept of Mendelian inheritance laws, including Mendel's second law of segregation.

[Generated by AI]

The ratios shown in these three crosses are classical Mendelian ratios that you should remember. They follow Mendel's second law of segregation.

Student view

Overview (/study/ap 422-cid-755105/o

Nature of Science

Aspect: Experiments

Theodor Boveri, an embryologist, conducted experiments on a roundworm, *Parascaris equorum,* which had large clear cells and only two pairs of chromosomes. He was able to follow the fate of certain chromosomes in individual cell lines very carefully and established that chromosomes in cells that produce gametes provide the continuity between generations.

Walter Sutton, another scientist, continued the work of Boveri and showed that he could distinguish 11 pairs of matched chromosomes taken from cells of the testes of a grasshopper, *Brachystola magna*, during meiosis. He observed the separation of the chromosomes during meiosis and stated that they 'may constitute the physical basis of the Mendelian law of heredity'.

The work of the two scientists, which identifies chromosomes (https://en.wikipedia.org/wiki/Chromosome) as the carriers of genetic material (https://en.wikipedia.org/wiki/Genes), is called the Boveri—Sutton chromosome theory.

Try the activity below to test your understanding of the terminology around Mendelian inheritance and independent assortment.

Activity

- **IB learner profile attribute:** Thinker
- **Approaches to learning:** Communication skills — Using terminology, symbols and communication conventions consistently and correctly
- **Time required to complete activity:** 20 minutes
- **Activity type:** Pair activity

Your teacher has printed out the Tarsia (https://drive.google.com/file/d/12FVAYHIXKs6iwnym15cQo9NwmIPEA9ha/view?usp=sharing) for you. In pairs, cut along all the solid lines to make nine triangles. Work as a team to pair up the correct statements to make one large triangle. You could do a timed challenge with other members of your class to see who can solve it correctly. Your pair can then meet with another couple to see whether you have the same combinations. If not, why not? Take turns to explain the reasoning behind your decisions and check with each other for understanding.

5 section questions

D3. Continuity and change: Organisms / D3.2 Inheritance

Linked genes (HL)

D3.2.18: Loci of human genes and the polypeptide products (HL) D3.2.19: Autosomal gene linkage (HL)

D3.2.20: Recombinants in crosses involving two linked or unlinked genes (HL) D3.2.21: Use of a chi-squared test on data from dihybrid crosses (HL)

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Higher level (HL)

Learning outcomes

By the end of this section you should be able to:

- Explain why linked genes fail to assort independently.
- Deduce genotypic and phenotypic possibilities of crosses of individuals heterozygous for two traits with those homozygous recessive for both traits in both linked and unlinked genes.
- Calculate statistical significance of observed vs calculated data using chi-squared tests.

Gene loci

Individuals with a family history of certain genetic diseases may wish to be tested to see whether they have the particular form of the gene responsible for the condition. Genetic counselling is a field of medicine in which highly skilled professionals interpret genetic data and explain it to patients and their families. Support is then given as they make decisions based on this information.

It might seem an impossible task to search through an individual's DNA for a single gene, but even though the human genome has 23 chromosomes with approximately 30 000 genes on those chromosomes, there are many that have been identified to specific locations. The genes within a human population are located in the same position called loci (singular locus) on specific chromosomes. Due to the huge number of genes, scientists have come up with codes to identify them; often letters, sometimes combined with numbers. Chromosomes have a short arm (p for *petit*) and a long arm (q). One of the genes that correlates to a susceptibility for breast cancer, *BRCA1*, can be found on the long (q) arm of chromosome 17. The *BRCA1* gene is in the same locus for all humans.

Each gene with its specific locus encodes for a polypeptide product. Use this website (https://www.ncbi.nlm.nih.gov/books/NBK22266/#A274) to explore the different chromosomes and some of the genes found on them. Look for the name of the gene (letters and numbers) and the condition that might arise if carrying the gene.

To find out more information about specific genes, search the database here (https://www.ncbi.nlm.nih.gov/gene/). You can enter either the gene name or the name of the protein it codes for. For example, to find the location of the genes associated with Huntington's disease, you can put 'Huntington's disease' in the search bar and it will bring up details of the *HTT*/huntingtin gene that results in the neurodegenerative disorder. If you scroll to the Genomic Context, you can see the precise location of this gene: 4p16.3 (Chromosome 4, p arm, location 16.3).

Linked genes

As mentioned previously, genes that are on the same chromosome are known as linked genes. It was Thomas Morgan in 1911 who first noticed that 'linked' traits would occasionally separate while other traits on the same chromosome would not. He was the one to propose that 'crossing over' between paired chromosomes could happen.

Theory of Knowledge

Thomas Morgan challenged the established view of Mendel's theories of inheritance and, by doing so, developed our understanding of gene linkage. To what extent is it important to challenge views that have been considered dependable?

Student view

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In your work, these linked genes should be shown in the format illustrated in **Figure 1**.

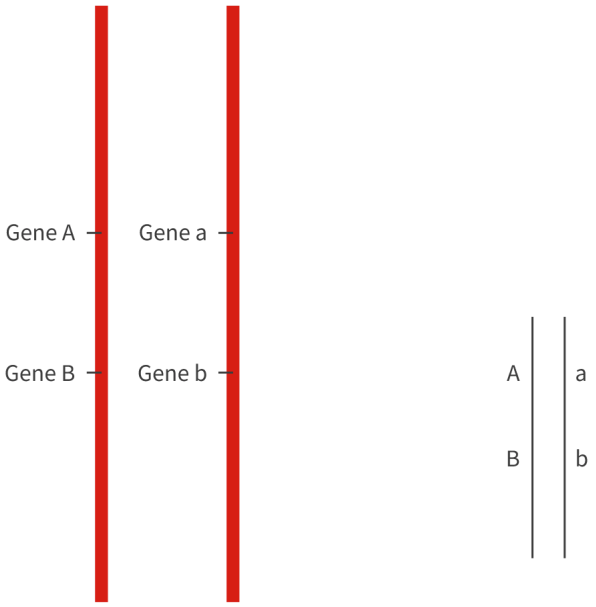

Figure 1. Linked genes on a pair of homologous chromosomes (left). The notation for representing linked genes is shown on the right.

More information for figure 1

The image is a diagram illustrating linked genes on a pair of homologous chromosomes. On the left side, there are two vertical, parallel red bars representing chromosomes. The left chromosome is labeled with 'Gene A' and 'Gene B' at different locations along its length, while the right chromosome is labeled 'Gene a' and 'Gene b' at corresponding locations. There are lines connecting Gene A with Gene a and Gene B with Gene b to indicate linkage.

On the right side, there is a notation showing the representation of these linked genes. It involves two short vertical lines labeled at the top with 'A' and 'a', and at the bottom with 'B' and 'b'. The notation exemplifies how linked genes are visually represented.

[Generated by AI]

Linked genes can fail to assort independently and so do not follow Mendel's laws. This may be because they are close to each other on the chromosome and therefore the genes travel together as a unit. However, if the genes are further apart on the chromosome, then there may be some exchange of material between the paternal and maternal chromosomes. This happens when the non-sister chromatids come close to each other in prophase I of meiosis during a process called synapsis. The corresponding parts of the non-sister chromatids might break and then reattach to the sister chromatid from the other parent. This is called crossing over and gives rise to more genetic variation. We can look at an example of this in **Figure 2**. Note that the point at which the chromatids are joined during crossing over is called the chiasma (plural, chiasmata).

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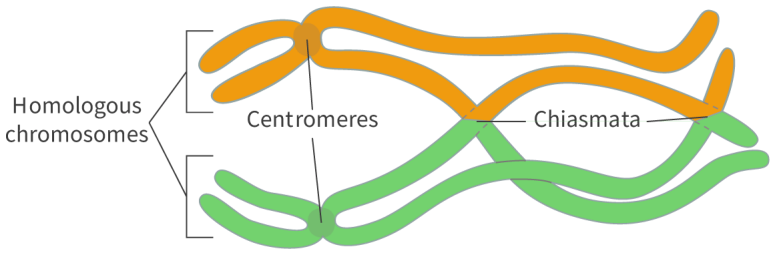

Figure 2. Crossing over during prophase I.

More information for figure 2

The diagram illustrates crossing over during prophase I of meiosis. It shows a pair of homologous chromosomes with one set in orange and one in green. The diagram highlights the key components, including centromeres and chiasmata, where crossing over occurs. Each chromosome is depicted with two chromatids. The centromeres are labeled in the middle of the chromosomes, indicating the point where the chromatids are joined. At the crossing sites, called chiasmata, there is an exchange of genetic material between non-sister chromatids of homologous chromosomes, represented by the intertwining of orange and green chromatids. The process is essential for increasing genetic variation through recombination.

[Generated by AI]

It is not guaranteed that crossing over will happen, nor that it will only happen once! You can imagine how the genetic variation can increase in meiosis using this manner of exchanging paternal and maternal genetic information in all 23 pairs of human chromosomes.

With linked genes, a question that we should ask is 'how close are the genes to each other?' If they are very close, then they are likely to be inherited together. The further apart they are from one another, the more likely it is that crossing over happens at least once. The distance between genes can be calculated based on the number of crossing overs. The further apart the genes are, the more crossing over episodes will likely have occurred. As the gametes are made in meiosis II, the haploid cells *may* contain the same combination of genes as the parent or, if crossing over has taken place, they may have a combination not found in either of the parents. Rather, it is a *re*combination of both of the parents' chromosomes. The frequency of recombinant genes is greater when the genes are further apart.

We can see this happening in **Figure 3** with an individual heterozygous for both genes and an individual homozygous recessive for both genes. We will initially observe what happens when there is no crossing over.

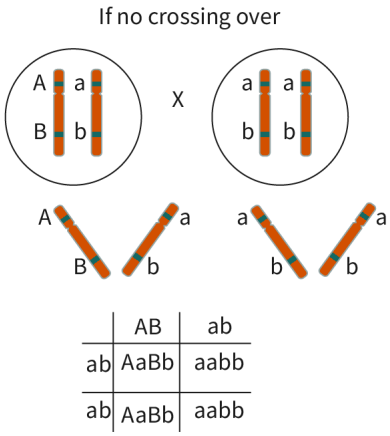

Figure 3. Dihybrid inheritance of linked genes with no crossing over.

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view

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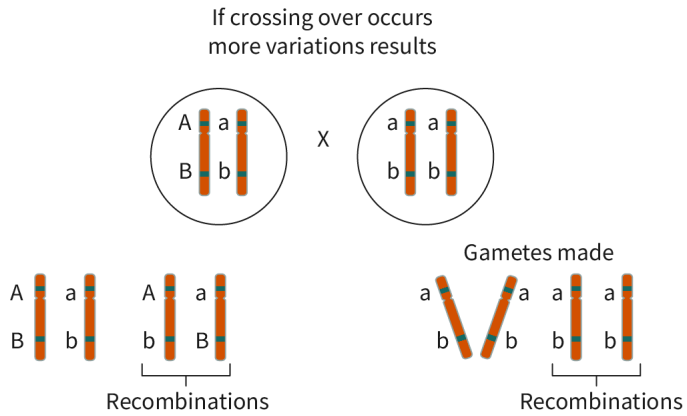
More information for figure 3

The diagram depicts dihybrid inheritance where no crossing over occurs between genes. At the top, two circles represent gametes from a heterozygous individual (with chromosomes labeled A, B) and a homozygous recessive individual (with chromosomes labeled a, b). There are no interactions indicated between them (e.g., no crossing over). Below the gametes, four chromosomes show the possible arrangements: two with AB and two with ab. At the bottom, a Punnett square shows the offspring results. Parental combinations are AB and ab (heterozygous) and ab and ab (homozygous recessive). The resulting genotypes in the square are AaBb and aabb, with homozygous offspring being present twice as frequently.

[Generated by AI]

We can see that the parental gametes that can form are AB and ab from the heterozygous parent and ab and ab from the homozygous recessive parent. The offspring can therefore only be of two types: AaBb or aabb.

However, if the genes are far enough apart to undergo crossing over and can assort independently, then we suddenly have many more gamete possibilities. This is shown in **Figure 4**.



	AB	ab	Ab	aB
ab	AaBb	aabb	Aabb	aaBb
ab	AaBb	aabb	Aabb	aaBb
ab	AaBb	aabb	Aabb	aaBb
ab	AaBb	aabb	Aabb	aaBb

Figure 4. Dihybrid inheritance of linked genes with crossing over.

More information for figure 4

The diagram illustrates dihybrid inheritance involving linked genes with crossing over. At the top of the image, two sets of chromosomes are shown, one labeled with alleles 'A' and 'B', and the other with 'a' and 'b'. These chromosomes illustrate the potential crossover points that can lead to genetic recombination.

Below this, two recombination scenarios are depicted. On the left, chromosomes are shown in a linear representation with 'A', 'a', 'B', and 'b' with a bracket indicating recombination. On the right, another set of chromosomes is shown with potential new gametes labeled 'a', 'b', 'A', and 'B', highlighting additional cross-over recombinations.

At the bottom, a Punnett square is used to display possible genetic outcomes based on these recombinations. The square is a 4x4 grid with columns labeled 'AB', 'ab', 'Ab', 'aB' and rows labeled 'ab'. Inside the grid, various combinations of the alleles are displayed such as 'AaBb', 'aabb', 'Aabb', and 'aaBb', showing all potential genetic outcomes after crossing over.

[Generated by AI]

Student view

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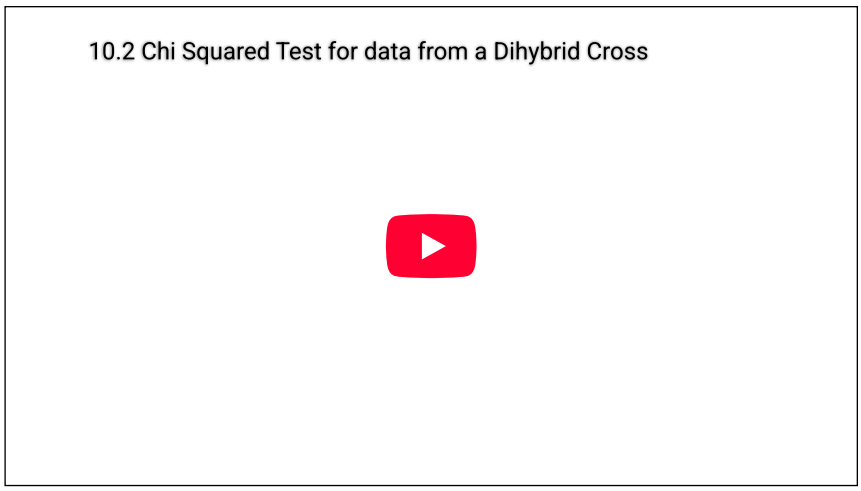
The gametes could now not only be the parental types AB and ab but could also be Ab and aB which are the recombinant gametes.

When crossing over with the ab gametes from the homozygous recessive individual, we now have potential offspring of AaBb; aabb; Aabb and aaBb in a 1:1:1:1 ratio.

Chi-squared test

In dihybrid crosses of linked genes, the predicted numbers of offspring of each phenotype produced can be calculated. However, in real life, these numbers might not be exact, but should be close to the expected ratio. A statistical test called the chi-squared test can be applied to analyse the statistical significance of the observed (real) numbers seen versus the expected (predicted) numbers. This will determine whether there is a significant association between the observed and expected data.

Let us take the rabbits produced in section D3.2.16–17 (/study/app/bio/sid-422-cid-755105/book/unlinked-genes-hl-id-45749/) that had the 9:3:3:1 ratio. This ratio is what would be expected if the genes were unlinked and segregated independently. If the genes *are* linked, we would expect that these ratios would not be seen. If our results do not follow the unlinked ratios, we can assume that the genes are linked and have not undergone independent assortment. The following video (**Video 1**) will help to explain how to carry out a chi-squared test.



Video 1. Chi-squared test for a dihybrid cross.

Before you set up the chi squared test, there is some information you need to gather. First of all, state your null hypothesis and alternative hypothesis.

The null hypothesis says that there is no statistically significant difference between the observed and expected data, that the data are due to chance and that the traits are unlinked and assort independently.

The alternative hypothesis says that there is a significant difference between the observed and expected data, suggesting that the genes are linked and that they do not assort independently.

Worked example 1: chi-squared test

Using the information from **Figure 5**, copy and complete the downloadable table below to determine whether or not we accept the null hypothesis stated above.

Out of 210 rabbits we are told that 120 are grey haired and black eyed; 40 are grey haired and red eyed; 37 are white haired and black eyed; 13 are white haired and red eyed.

Student view

Female rabbit \ Male rabbit	GB	Gb	gB	gb
GB	GGBB	GGBb	GgBB	GgBb
Gb	GGBb	GGbb	GgBb	Ggbb
gB	GgBB	GgBb	ggBB	ggBb
gb	GgBb	Ggbb	ggBb	ggbb

	Grey haired; black eyed	9
	Grey haired; red eyed	3
	White haired; black eyed	3
	White haired; red eyed	1

Figure 5. Dihybrid cross.

More information for figure 5

The image shows a Punnett square representing a dihybrid cross between rabbits, considering hair color and eye color. The top row is labeled "Male rabbit" with columns for each genotype: GB, GB, Gb, and Gb. The left column is labeled "Female rabbit" with rows for each genotype: GB, Gb, gB, and gb. The grid is filled with genotypes resulting from the cross, such as GGBB, GGBb, GgBB, and GgBb. Below the Punnett square is a legend indicating phenotypic ratios: blue for grey haired and black eyed (9), green for grey haired and red eyed (3), purple for white haired and black eyed (3), and red for white haired and red eyed (1).

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Table 1 (https://d3vrb2m3yrmyfi.cloudfront.net/media/edusys_2/content_uploads/Biology D3.2.18-21 WE1.5696b7b1001e0e9b3493.pdf)

Solution steps	Calculations
Step 1: Put in the observed frequencies (see row 1 of the table).	Values taken from the data provided.
Step 2: Calculate the expected frequencies. These can be calculated from the 9:3:3:1 ratio as if there is independent assortment (see row 2).	Calculate the values by using the expected ratio of, for example, $\frac{9}{16} \times \text{total number of individuals}$: $\frac{9}{16} \times 210 = 118.100$
Step 3: Next the observed (O) — expected (E) values are calculated (see row 3).	e.g. $180.000 - 118.100 = 1.900$
Step 4: Next square these values $(O - E)^2$ (row 4).	e.g. $(1.900)^2 = 3.610$
Step 5: This value is then divided by the expected value (row 5).	e.g. $\frac{3.610}{118.100} = 0.03$
Step 6: The sum of all these is calculated and this value is χ^2 (see row 6).	e.g. 0.186

Solution steps	Calculations
Step 7: Calculate the degrees of freedom (df).	This is (number of rows — 1) × (number of columns — 1). In this case, this is $(2-1)\times(4-1)=3$.
Step 8: The final step is to use a chi-squared distribution table and look at the column where 95% confidence interval or 0.05 intersects with the row corresponding to 3 df (see **Table 2**).	
Step 9: Determine whether the χ^2 value calculated is less than the critical value from the table.	The χ^2 value calculated of 0.186 is less than the critical value of 7.815 from the table and so the null hypothesis is accepted.

Table 1 (solution). Data calculated for the chi-squared worked example.

	Grey haired, black eyed	Grey haired; red eyed	White haired; black eyed	White hai red eye
Observed	120.00	40.000	37.000	13.00(
Expected	$\frac{9}{16}\times 210=118.100$	$\frac{3}{16}\times 210=39.400$	$\frac{3}{16}\times 210=39.400$	$\frac{1}{16}\times 210=1$
$\mathrm{O}-\mathrm{E}$	1.900	0.600	−2.400	−0.10(
$(\mathrm{O}-\mathrm{E})^2$	3.610	0.360	5.760	0.010
$\frac{(\mathrm{O}-\mathrm{E})^2}{\mathrm{E}}$	0.030	0.009	0.146	0.001
$X^2=\sum\frac{(\mathrm{O}-\mathrm{E})^2}{\mathrm{E}}$				

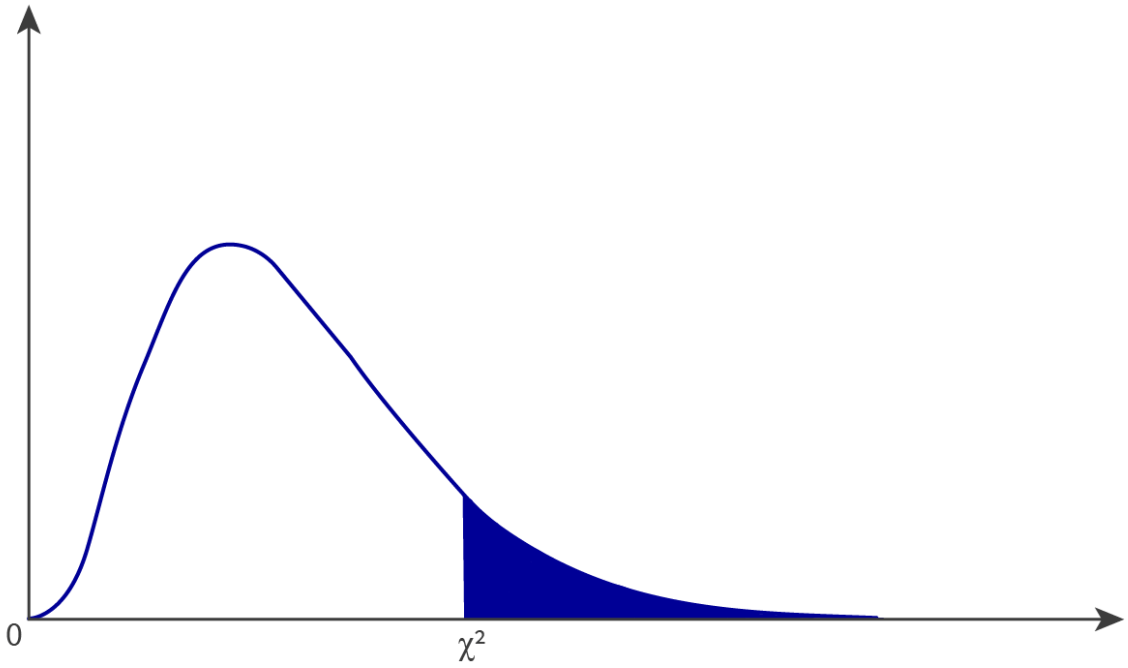

Figure 6. The shaded area is equal to α for $\chi^2=\chi^2\alpha$.

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Table 2. A distribution table for the chi-squared test.

df	$\chi^2_{.995}$	$\chi^2_{.990}$	$\chi^2_{.975}$	$\chi^2_{.950}$	$\chi^2_{.900}$	$\chi^2_{.100}$	$\chi^2_{.050}$	$\chi^2_{.025}$	$\chi^2_{.010}$	$\chi^2_{.005}$
1	0.000	0.000	0.001	0.004	0.016	2.706	3.841	5.024	6.635	7.879
2	0.010	0.020	0.051	0.103	0.211	4.605	5.991	7.378	9.210	10.597
3	0.072	0.115	0.216	0.352	0.584	6.251	7.815	9.348	11.345	12.838
4	0.207	0.297	0.484	0.711	1.064	7.779	9.488	11.143	13.277	14.860
5	0.412	0.554	0.831	1.145	1.610	9.236	11.070	12.833	15.086	16.750

The null hypothesis is therefore accepted and we can conclude that there is no significant difference between the observed and expected values. As a result, we can assume that the genes are unlinked and do segregate independently.

Try the activity below to discover the genetics of corn.

Activity

- **IB learner profile attribute:** Inquirer
- **Approaches to learning:** Thinking skills — Reflecting on the credibility of results
- **Time required to complete activity:** 45 minutes
- **Activity type:** Pair activity

In this activity, you will work in pairs to study the genetics of some corn.

Look at **Figure 7** and then download the worksheet using the button below. With your classmate, you will examine a photo of an ear of corn and determine the type of cross and genes responsible for the colouration and texture of some of the corn kernels shown. Consider the following four grain phenotypes: purple and smooth (A), purple and shrunken (B), yellow and smooth (C), yellow and shrunken (D).

Worksheet (https://d3vrb2m3yrmyfi.cloudfront.net/media/edusys_2/content_uploads/Biology D3.2.18-21 ACTIVITY.cd7f723031ea611dab06.pdf)

Figure 7. Corn kernels with different colours and textures.
Source: "CSIRO ScienceImage 3498

Student view

Summary and key terms

- Eukaryotic organisms have a sexual life cycle with a pattern of inheritance that combines unique gametes from the male and female allowing for increased variation in a population.
- Mendel was the first to describe a unit of inheritance (what we now call a gene) and was able to predict the outcomes of certain breeding events.
- Inheritance of one gene causing a single trait is known as monohybrid inheritance, two genes influencing two traits as dihybrid inheritance and when many genes are involved in a trait it is called polygenic inheritance.
- Different forms of the gene are called alleles and can be dominant or recessive, codominant or incompletely dominant. The combination of alleles forms the genotype. This will then dictate the phenotype of the individual including dominant and recessive diseases.
- Phenotype is changeable (plastic) and can be altered by the impact of environmental conditions.
- There may be many different alleles for a gene. If there are more than two alleles it is known as multiple alleles. Changes of single nucleotides that are seen across a population are known as single nucleotide polymorphisms (SNPs).
- Sex is determined by the chromosome that is inherited from the father. Certain genes are present on the X chromosome and so males are more predisposed to certain X-linked conditions.
- Pedigree charts are used to visualise inheritance patterns in families.

Higher level (HL)

- Whether the genes are linked (on the same chromosome) or unlinked (on different chromosomes or far enough apart on the same chromosome) will determine their ability to separate and reassort in meiosis to contribute to the gametes.
- Chi-squared tests can be used to examine the statistical significance of the observed values of offspring when compared with calculated expected values.

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Key terms

Review these key terms. Do you know them all? Fill in as many gaps as you can using the terms in this list.

1. _____ are lengths of DNA and are the basic unit of heredity. Many of them encode for _____.
2. _____ are different forms of a gene and can be _____ or dominant.
3. The _____ describes the combination of alleles, whereas the _____ describes the outward expression of the genotype.
4. Variation in a population is increased by a _____ life cycle as the gametes produced by the cell division of _____ are unique.
5. The pattern of inheritance throughout a family can be shown diagrammatically using a _____.
6. [HL] Genes can be on the same chromosome – _____ – or on different chromosomes – _____.
7. [HL] When alleles in one or more traits are sorted into gametes independently of each other, this is known as _____.

linked | genotype | proteins | phenotype | sexual | Alleles | recessive | Genes | meiosis | pedigree chart | unlinked | independent assortment

Check

Interactive 1. Genetic Inheritance and Variation.

Checklist

What you should know

After studying this subtopic you should be able to:

- Outline that haploid cells (with a single copy of a gene) produced by each parent can fuse to form a diploid zygote with two copies of a gene.
- Explain methods for how flowering plants are genetically crossed.
- Distinguish between genotype (combination of inherited alleles) and phenotype (observable traits resulting from genotype plus environmental factors).
- Explain the ways that members of the same species can have variety in the gene pool.
- Describe the inheritance of different blood types.

Student view

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- Compare and contrast the differences between incomplete dominance and codominance.
- Describe that the sperm determines sex in humans.
- Describe haemophilia as an example of a sex-linked genetic disorder.
- Illustrate how pedigree charts are used to determine inheritance in family members.
- Distinguish between continuous variation such as skin colour and discrete variation such as ABO blood group.
- Illustrate continuous variables using box-and-whisker plots.

Higher level (HL)

- Explain how unlinked genes segregate and assort independently in meiosis.
- Predict the inheritance of pairs of unlinked genes in dihybrid crosses.
- Predict genotypic and phenotypic ratios in dihybrid crosses of unlinked autosomal genes using Punnett grids.
- Explain why linked genes fail to assort independently.
- Deduce genotypic and phenotypic possibilities of crosses of individuals heterozygous for two traits with those homozygous recessive for both traits in both linked and unlinked genes.
- Calculate statistical significance of observed vs calculated data using chi-squared tests.

Investigation

Section Student... (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/investigation-id-46540/print/) Assign

- **IB learner profile attribute:** Inquirer
- **Approaches to learning:** Thinking skills – Providing a reasoned argument to support conclusions
- **Time required to complete activity:** 30 minutes
- **Activity type:** Individual activity

Analysing a specific pedigree chart to determine the inheritance pattern involved

As discussed in section D3.2.11–13 (/study/app/bio/sid-422-cid-755105/book/genetics-of-the-y-chromosome-id-46206/), investigating pedigree charts can provide insights into the inheritance patterns of specific traits or genetic disorders.

In this activity you will study the inheritance pattern of a specific genetic disorder within a family by analysing a pedigree chart and determining the possible genotypes of family members.

Section Student... (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/summary-and-key-terms-id-46539/print/) Assign

~~Medical~~ history and information on the genetic disorder

Galactosaemia is an inherited metabolic disorder that prevents the body from breaking down the sugar galactose. This causes galactose to build up in the blood to toxic levels, which can lead to serious complications such as an enlarged liver, kidney failure, cataracts in the eyes or brain damage. People with the disorder have to avoid dairy products, breast milk and most baby formulas. Galactosaemia can be life-threatening to infants if they are not diagnosed quickly. With early diagnosis and a lactose-restricted diet, people with galactosaemia can go on to lead relatively normal lives.

Student view

Figure 1 shows the pedigree chart of the family under study.

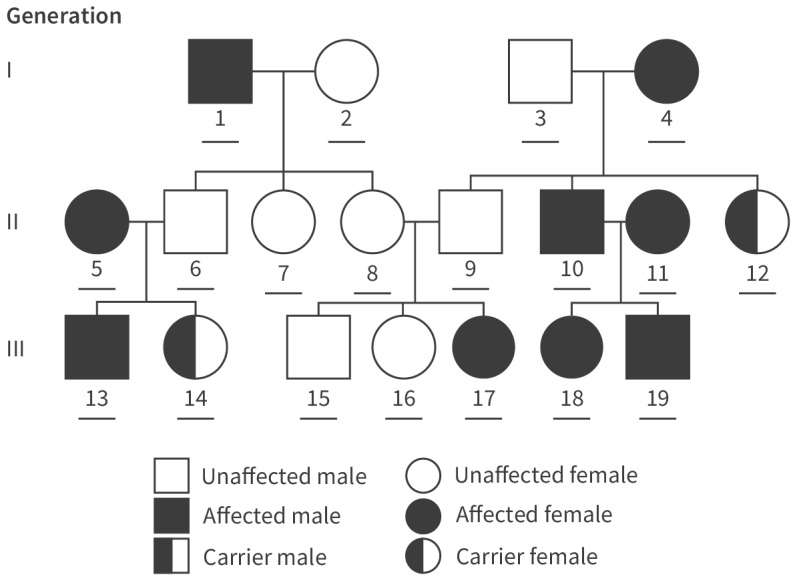

Figure 1. Pedigree chart for family with galactosaemia.

More information for figure 1

The image is a pedigree chart illustrating family generations and the inheritance pattern of galactosaemia. It is organized into three generations, labeled I, II, and III, with connecting lines indicating familial relationships. There are various symbols representing individuals: squares for males and circles for females. The chart uses different shadings to denote genetic attributes: solid fills for affected males and females, half fills for carrier males and females, and empty shapes for unaffected individuals. Each individual is numbered for reference.

In Generation I, individuals 1 and 2 are shown as parents, with individual 1 being an affected male and individual 2 an unaffected female. They have four children in Generation II: an affected female (5), an unaffected male (6), unaffected females (7 and 8). Another couple in Generation I, comprising an unaffected male (3) and an affected female (4), have six children in Generation II: unaffected and affected males and females (9 to 12).

Generation II individuals 5 and 6 have one child in Generation III (13), who is an affected male, and a carrier female (14). Individuals 9, 10, and 11, along with their partners, have children 15 to 19 in Generation III, showing various combinations of affected, carrier, and unaffected statuses.

[Generated by AI]

Your task

1. Examine the detailed pedigree chart of a family exhibiting the genetic disorder galactosaemia (**Figure 1**). The chart includes three generations (I, II, III) of affected, unaffected and carrier individuals. For some of the family members, it is not known if they are unaffected or carrier individuals, and so the symbols have been left deliberately empty. However, we do know that these individuals do not have the disorder themselves.
2. Analyse the pedigree chart to identify patterns of inheritance. Look for clues such as affected individuals in multiple generations and the presence of unaffected carriers.
3. Using information gathered from the pedigree chart, determine if the disorder would be autosomal or sex-linked.
4. From examining the offspring of parents numbered 8 and 9, identify if the inheritance of the disorder is recessive or dominant.
5. Determine the possible genotypes of family members (1–19) based on the identified inheritance pattern. Record your findings in a table similar to **Table 1**.

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Table 1. Example results table.

Generation	Individual no.	Genotype(s)

6. If there is a possibility that an individual could be two genotypes, list both, with the most probable one first.
7. Construct a hypothetical pedigree chart using any two individuals from the third generation (III) and the predicted genotypes to confirm the consistency of the proposed inheritance pattern.
8. Compare your constructed pedigree with the original pedigree chart to validate the accuracy of the predicted genotypes and inheritance pattern.
9. Discuss any limitations or inconsistencies in the pedigree chart that could affect the interpretation of the inheritance pattern.

Analysis

1. Evaluate the consistency of the observed pedigree with the expected pattern of inheritance that you have identified.
2. Compare the predicted genotypes with the observed phenotypes in the family members.
3. Assess any complexities or deviations from the typical identified inheritance pattern and discuss the possible reasons for these discrepancies.

Conclusion

Summarise the findings of the investigation and discuss the extent to which the pedigree chart analysis supports your analysis regarding the identified inheritance pattern. Reflect on the significance of understanding genetic inheritance patterns for diagnosing and managing genetic disorders within families. Also, consider potential implications for genetic counselling and family planning.

Reflection

Section Student... (0/0) Feedback Print (/study/app/bio/sid-422-cid-755105/book/reflection-id-46897/print/) Assign

Teacher instructions

The goal of this section is to encourage students to reflect on their learning and conceptual understanding of the subject at the end of this subtopic. It asks them to go back to the guiding questions posed at the start of the subtopic and assess how confident they now are in answering them. What have they learned, and what outstanding questions do they have? Are they able to see the bigger picture and the connections between the different topics?

Students can submit their reflections to you by clicking on 'Submit'. You will then see their answers in the 'Insights' part of the Kognity platform.

Student view

Reflection

Overview (/study/ap 422- cid- 755105/o

Now that you've completed this subtopic, let's come back to the guiding questions introduced in The big picture (/study/app/bio/sid-422-cid-755105/book/the-big-picture-id-43244/).

- What patterns of inheritance exist in plants and animals?
- What is the molecular basis of inheritance patterns?

With these questions in mind, take a moment to reflect on your learning so far and type your reflections into the space provided.

You can use the following questions to guide you:

- What main points have you learned from this subtopic?
- Is anything unclear? What questions do you still have?
- How confident do you feel in answering the guiding questions?
- What connections do you see between this subtopic and other parts of the course?

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