AQA Biology GCSE

Genetics: Inheritance

DNA and the Genome

Key Words:

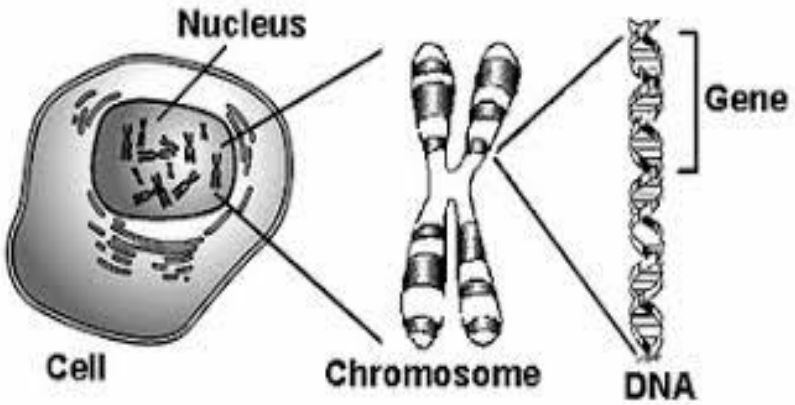
Polymer	A large molecule formed from many identical smaller molecules known as monomers.
DNA	DNA stands for deoxyribonucleic acid. DNA carries genetic information. It has all the instructions that a living organism needs to grow, reproduce and function.
Gene	A short section of DNA that codes for a protein
Genome	An organisms complete set of genetic information (in every cell)
Chromosomes	The structure that DNA is organised into – humans have 23 pairs of chromosomes in the nucleus
Nucleus	The nucleus controls what happens inside the cell. Chromosomes are structures found in the nucleus of most cells. The plural of nucleus is nuclei.
Double helix	The shape of the DNA molecule which has two strands twisted together in a spiral
Genetic Code	The code formed by the order of the bases in DNA that determines an organism's characteristics.

Part 1: State the structure and role of DNA

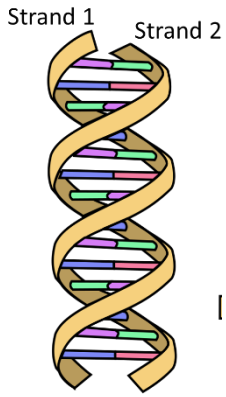
Information: *Read the following notes*

DNA is a **polymer** made up of **two strands** forming a **double helix** *('Double' = two strands 'Helix' = twisted around each other*).

The two strands are held together by chemicals called 'bases'. DNA is a chemical which contains genetic information or the "code for life", small sections of the DNA called **genes** code for a particular sequence of amino acids in order to make a specific protein.

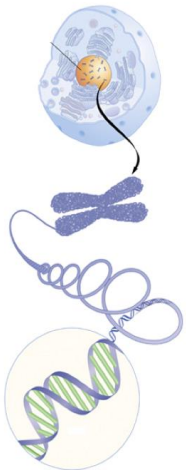


An organisms complete set of genetic information (in every cell) is called its **genome.**



Part 2: Describe how genetic material is organised.

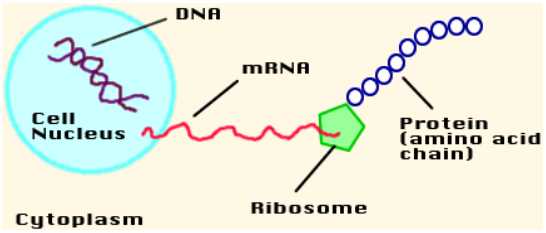
Information: *Read the following notes*



DNA is coiled into structures called **chromosomes.** The chromosomes are found in the **nucleus** of each cell. Human body cells contain 23 pairs (46) chromosomes

A gene is a short section of DNA that codes for a specific protein. The code of each gene tells the ribosomes the order to place certain amino acids.

We say each gene 'codes for' a specific protein because if you change the order of the amino acids a different protein would be made.



Recall questions: *Answer these in your book, use the information above to help you*

1. The genetic material in the nucleus of a cell is….
2. Describe the structure of DNA.
3. Name the structure within which DNA is contained.
4. What is a gene?
5. What is the function of a gene?
6. What is a genome?
7. What is a change in the DNA sequence called?
8. What are chromosomes?
9. How many chromosomes are in one human body cell?

Part 3: Discuss the importance of understanding the human genome.

The Human Genome Project (HGP) was an international scientific research project with the goal of determining the sequence of chemical bases which make up all of human DNA which is over 3 billion bases long!

The importance of identifying and knowing the order of genes in the human genome is so that we can:

- **To search for genes linked to different types of disease**
- **To understand and treat inherited disorders**
- **To trace early human migration patterns**

Recall questions: *Answer these in your book, use the information above to help you*

10. State 3 ways in which understanding of the human genome will be important.

Genotype and Phenotype

Key Words:

Alleles	Different form of the same gene
Characteristics	A **characteristic** is an observable (can see it or measure it) quality, trait or feature of an individual e.g. hair colour, eye colour, flower colour, leaf shape.
Dominant	An allele that always expresses itself whether it is it is partnered by a recessive allele or by another like itself.
Recessive	Describes the variant of a gene for a characteristic which is masked or suppressed in the presence of the dominant variant. A recessive gene will remain dormant unless it is paired with another recessive gene.
Genotype	The alleles that an organism has for a particular characteristic, usually written as letters.
Phenotype	The visible characteristics of an organism which occur as a result of its genes.

Part 1: Define the terms genotype and phenotype.

Information: *Read the following notes*

When we describe how different organisms appear or look, we are describing their **phenotype –** their observable characteristics. A **characteristic** is an observable (can see it or measure it) quality, trait or feature of an individual e.g hair colour, eye colour, flower colour, leaf shape.

Colour phenotypes: white, pink and red

Different forms of the same gene are known as **alleles**. An organism's **genotype** is the alleles an individual has for a certain characteristic.

Recall questions: *Answer these in your book, use the information above to help you*

1. What is an allele?
2. What is a genotype?
3. What is a phenotype?

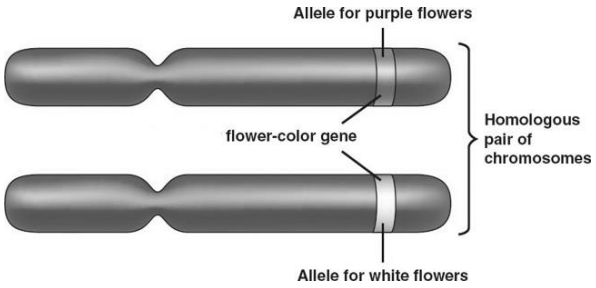
Part 2: Describe how some characteristics are controlled by a certain allele.

Information: *Read the following notes*

There are two copies of every chromosome in a body cell nucleus (one copy inherited from the mother the other copy inherited from the father).

Therefore, there are two copies of every gene. These may have different alleles and the combination of the two alleles (the **genotype**) determines the characteristic (the **phenotype**).

In this example there is a gene for flower colour, this plant has two copies of the flower colour gene one on each chromosome. This plant has two different alleles (versions of the flower colour gene) one allele codes for purple petals the other codes for white petals.



Recall questions: *Answer these in your book, use the information above to help you*

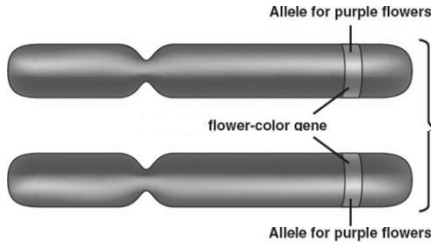
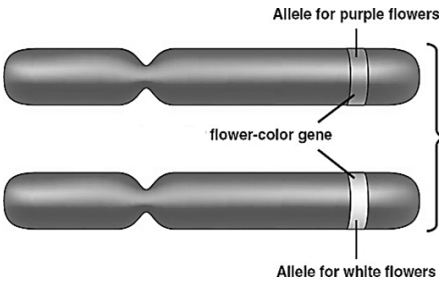
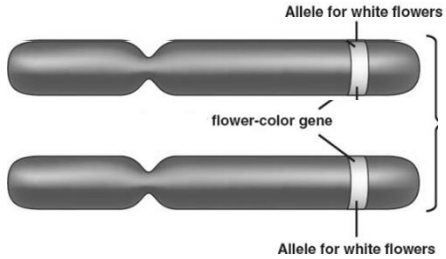
4. What percent of your chromosomes have you inherited from your father?
5. What is the word used to describe the alleles an organism has for a gene?
6. What word describes the physical characteristics an organism has?

Part 3: Explain how a dominant allele is always expressed

Information: *Read the following notes*

For most genes, one allele is said to be **dominant** and the other **recessive**. Only one dominant allele needs to be present for the dominant phenotype to be expressed. This means individuals that have two dominant alleles OR one dominant and one recessive allele will express the dominant phenotype. Two copies of recessive alleles need to be present for the recessive phenotype to be expressed. Due to this relationship, we often refer to the alleles using the same letter, the dominant in UPPER case and the recessive in lower case e.g. "B" is dominant, "b" is recessive.

For example: In the gene for flower colour purple is the dominant allele, so our code is P= purple and p=white

Chromosomes/Gene	Genotype	Phenotype
	PP Dominant Alleles Only	Purple Flowers
	Pp Dominant and Recessive Alleles	Purple Flowers
	pp Recessive Alleles only	White Flowers

Organisms which have two copies of a recessive allele and show the recessive phenotype cannot pass on any dominant alleles to their offspring unless they sexually reproduce with a mate that has dominant alleles. E.g. two plants with white flowers cannot produce plants with purple flowers but one white plant could reproduce with a purple plant and their offspring would have purple flowers. You can use this as evidence to tell if a characteristic is dominant or recessive!

Recall questions: *Answer these in your book, use the information above to help you*

7. What does the term "dominant" mean when used to describe an allele?
8. Give an example of a dominant genotype using the letter G.
9. What does the term "recessive" mean when used to describe an allele?
10. Give an example of a recessive genotype using the letter G.
11. How can you identify if a characteristic or phenotype is recessive?

Genetic Inheritance and Sex Determination

Key Words:

Heterozygous	This describes a genotype in which the two alleles for a characteristic are different.
Homozygous	This describes a genotype in which the two alleles for the characteristics are identical.
Punnet Square	A grid that shows the possible combinations of alleles that can result at fertilisation.

Part 1: Define the terms homozygous and heterozygous

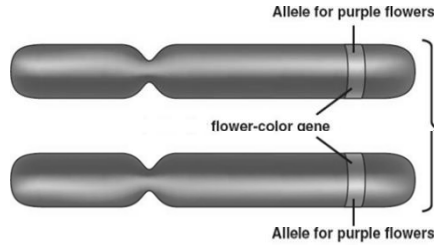
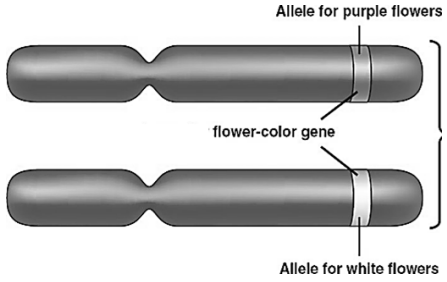
Information: *Read the following notes*

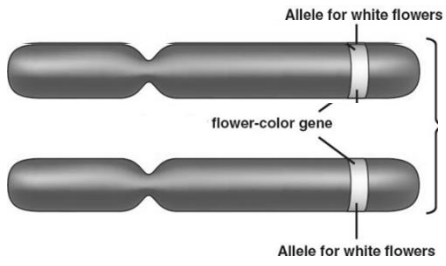
If the two alleles of a gene are the **same**, the individual is described as **homozygous. Example: BB or bb**

If two alleles of a gene are **different**, the individual is described as **heterozygous. Example: Bb**

Example: The gene for flower colour in pea plants has two alleles, purple and white.

The purple allele is represented by "P" and the white allele by "p".

Chromosomes/Gene	Genotype	Phenotype
	PP Dominant Alleles Only ***Homozygous dominant***	**The homozygous dominant phenotype is purple flowers**
	Pp Dominant and Recessive Alleles ***Heterozygous***	**The heterozygous phenotype purple flowers**

	pp Recessive Alleles Only ***Homozygous recessive***	**The homozygous recessive phenotype is white flowers**

Recall questions: *Answer these in your book, use the information above to help you*

1. What does the term "heterozygous" mean?
2. What does the term "homozygous" mean?

Part 2: Use Punnett squares to predict and interpret the results of a genetic cross.

Information: *Read the following notes*

A **Punnett square** can be used to perform a **genetic cross** to work out the **probability** of a certain allele being present in the offspring of two parents each time they reproduce. The information in a punnet square is labelled here.

	Parent 1 allele 1	Parent 1 allele 2
Parent 2 allele 1	Offspring alleles chance 1	Offspring alleles chance 2
Parent 2 allele 2	Offspring alleles chance 3	Offspring alleles chance 4

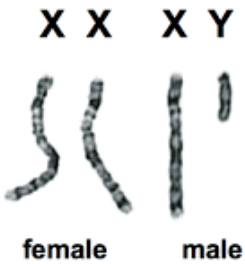
Remember - Probability means 'how likely' or 'what chance' - normally a fraction or a % example: ¼, 0.25 or 25%

	P	*p*
P	*PP* 25%	*Pp* 25%
p	*Pp* 25%	*pp* 25%

In this example using our Pp alleles for purple and white flowers again:

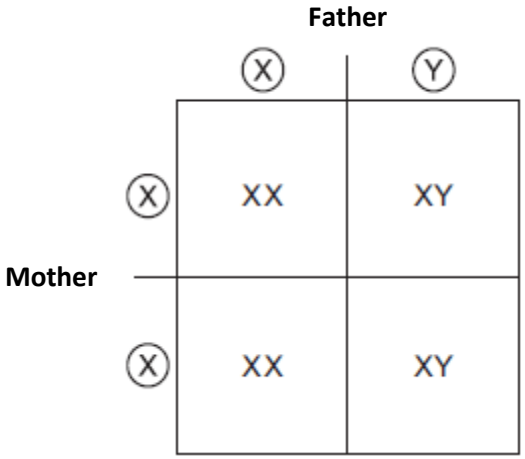
Genotypes
- ¼ or 25% chance of the homozygous dominant PP
- ½ or 50% chance (2x 25%) of the heterozygous Pp
- ¼ or 25% chance of the homozygous recessive

Phenotypes
- ¾ or 75% chance of having purple flowers (as it is dominant)
- ¼ or 25% chance of having white flowers (as it is recessive)



A sex chromosome is a type of chromosome that determines the sex of an organism. Humans and most mammals have two sex chromosomes, the X and the Y. Females have two X chromosomes in their cells, while males have one X and one Y chromosomes in their cells.

We can use a punnet square to show that the probability of getting a girl or a boy each time an embryo is fertilised is always 50%.



Recall questions: *Answer these in your book, use the information above to help you*

3. What is the genotype of a female?
4. What is the genotype of a male?

Inherited Disorders

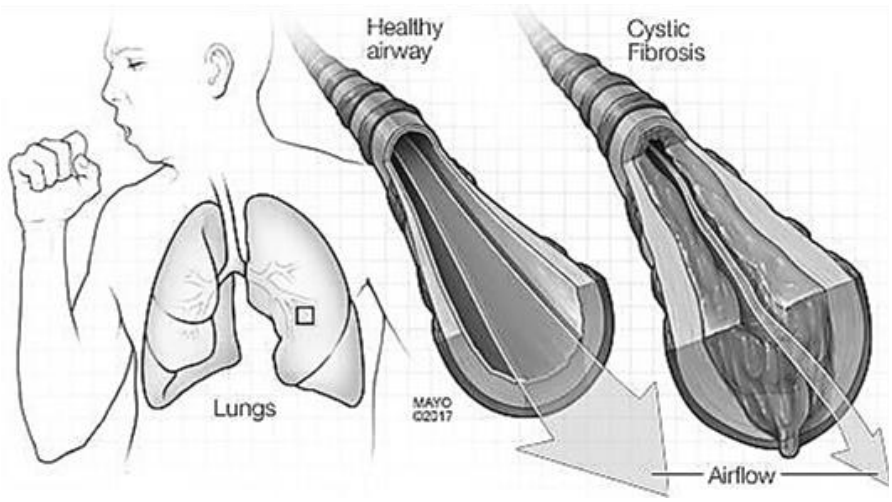
Key Words:

Polydactyly	Polydactyly is a genetic disorder caused by a dominant allele. People born with polydactyly have extra fingers or toes.
Cystic fibrosis	Cystic fibrosis is a genetic disorder caused by 2 recessive alleles.
Embryo	A ball of cells formed after fertilisation that undergoes mitosis to grow and form a foetus.
Mutation	A change to the DNA base sequence.

Part 1 - State (at least) two inherited disorders (cystic fibrosis and polydactyly)

Information: *Read the following notes*

Cystic Fibrosis is a genetic disorder caused by a mutation. It affects many organs including the: lungs, digestive system and reproductive system. It affects mucus made by cells in different areas of the body causing it to become very thick and sticky.

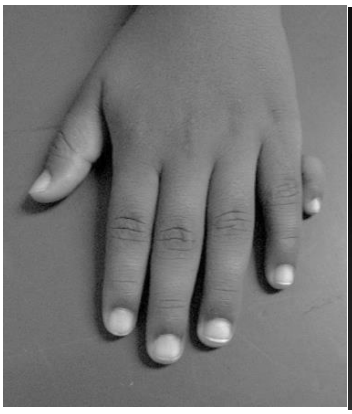


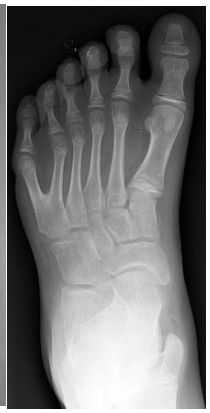
Organs such as the lungs can become clogged up, this makes breathing harder and increases their chance of getting chest infections due to build-up of dust and dirt. Their pancreatic duct can also be blocked, resulting in less digestive enzymes making their way into the small intestine. This can reduce the amount of nutrients they can get from their food – this often leads to them being underweight.

It cannot be cured as it is a genetic disorder, but it can be treated to help reduce symptoms. Physiotherapy can help keep area's clear of mucus and antibiotics are used to prevent infection. Later in life a lung transplant may be needed.

Cystic fibrosis is a recessive genetic disorder (only people who are homozygous recessive (have two copies of the recessive allele) for the cystic fibrosis gene have the disorder).

Not all genetic diseases are recessive.





Polydactyly is caused by a dominant allele. People born with polydactyly have extra fingers or toes.

This genetic disorder is not harmful. People can choose to have extra digits removed with surgery but some keep them.

Some dominant genetic disorders have a much more widespread effects on the way the body works and cause serious symptoms, they are often life limiting e.g. Huntington's disease.

Recall questions: *Answer these in your book, use the information above to help you*

1. What are the symptoms associated with cystic fibrosis?
2. What type of genetic disorder is cystic fibrosis?
3. How can cystic fibrosis be treated?
4. What is polydactyly?
5. Is polydactyly a dominant or recessive condition?
6. State the genotype(s) that would result in polydactyly.

Part 2: Describe the difference in inheritance patterns for dominant/recessive allele disorders.

Information: *Read the following notes*

Cystic fibrosis is caused by a recessive allele. This means that only homozygous recessive people are suffering from the disorder and heterozygous people are completely normal, but they have a chance of having a child which suffers from it. They are known as carriers.

To the right is the Punnett square for a cross between two heterozygous carriers. C= healthy, c= cystic fibrosis

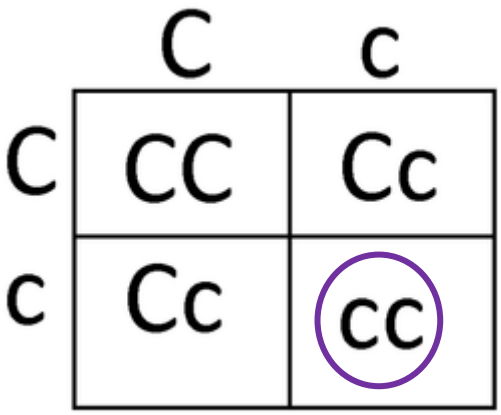
In this example:

Genotypes

- 25% chance of the homozygous dominant CC
- 50% chance of the heterozygous Cc
- 25% chance of the homozygous recessive cc

Phenotypes

- 25% of being unaffected CC
- 50% of having no symptoms but being a carrier Cc
- 25% chance of suffering from cystic fibrosis cc (circled)



Polydactyly is caused by a dominant allele. This means that only one parent needs to pass on the faulty allele for the child to suffer from polydactyly, So a homozygous dominant parent is guaranteed to have a polydactyly child and a heterozygous parents will have a chance of having one.

To the right is a Punnett square showing the chances of having child with polydactyly when one parent with the disorder mates with a homozygous recessive partner.

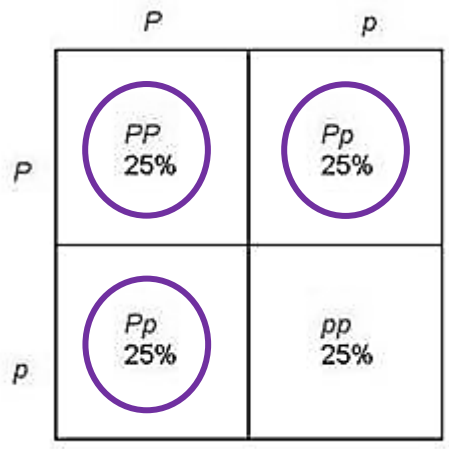
In this example:

Genotypes

- 25% chance of the homozygous dominant PP
- 50% chance of the heterozygous Pp
- 25% chance of the homozygous recessive pp

Phenotypes

- 75% chance of having polydactyly (circled)
- 25% chance of being unaffected



<u>Recall questions:</u> *Answer these in your book, use the information above to help you*

7. What is cystic fibrosis?
8. Is cystic fibrosis a dominant or recessive condition?
9. State the genotype(s) that would result in cystic fibrosis.

Part 3: Discuss the issues surrounding embryo screening.

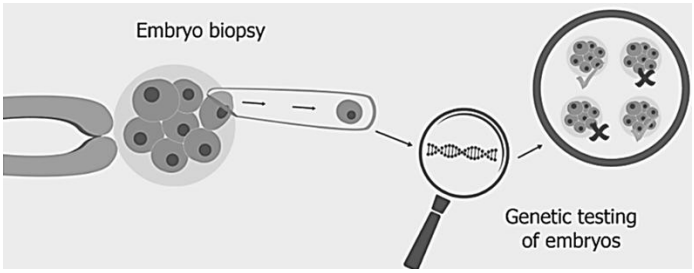
<u>Information:</u> *Read the following notes*

Genetic testing involves analysis of a person's DNA to see if they carry alleles that cause genetic disorders. It can be done at any stage in a person's life. If people know they are carriers of a genetic disorder and they would like to have

children but not pass on the alleles that cause their genetic disorder they can have their embryos screened before pregnancy occurs.

Pre-implantation genetic diagnosis (PGD) or **embryo screening** can be used on embryos before placing them into the uterus of the mother. Fertility drugs stimulate the release of several eggs. The eggs are collected and fertilised with sperm in a Petri dish. This is known as in vitro fertilisation (IVF).

Once the embryos have reached the eight-cell stage, one cell is removed from each. These cells are tested for the disorder causing alleles. Only embryos that do not contain the alleles are then implanted into the uterus, embryos with the alleles for the disorder are destroyed.



Testing can also be done during the pregnancy. This testing uses a needle to take a sample of the growing foetus's blood in the womb. It is offered to couples who may have an increased risk of producing a baby with an inherited disorder, but it cannot detect all the inherited disorders unlike embryo screening.

The **main advantage** of screening is it allows parents the ability to have healthy children without the disorder or they can make an informed choice to continue or terminate a pregnancy.

But there are also some **disadvantages**:

- Testing can cause miscarriage by damaging the embryo or cause harm to the mother or unborn foetus (if done during pregnancy)
- People can be against these tests for moral and religious reasons. Some people have a strong belief that it is **immoral or unethical** to **destroy embryos** or terminate foetuses which could have lived.
- The tests are often expensive, and the results are not always reliable

Recall questions: *Answer these in your book, use the information above to help you*

1. State the benefit of embryo screening.
2. State one concern of screening embryos.

Genetic Diagrams

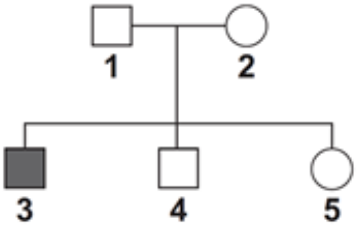
Key Words:

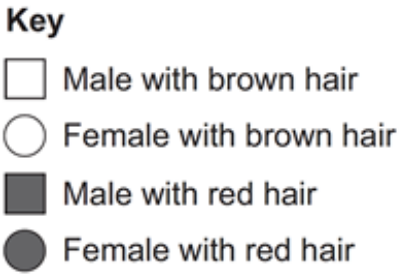
Genetic diagram/family tree/Pedigree Chart	A diagram which shows the inheritance of a genetic condition in a family across generations.

Part 1 - Identify alleles on a genetic diagram or 'family tree'

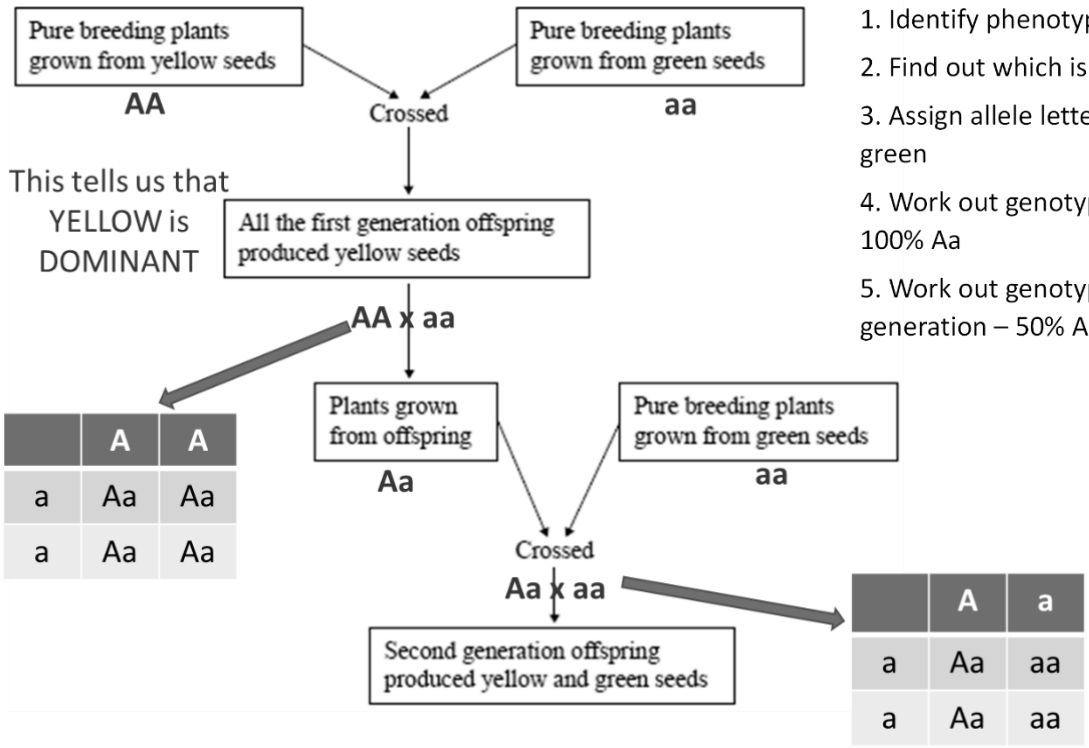
Information: *Read the following notes*

Genetic diagrams or family trees show how characteristics are inherited across multiple generations. They can be abstract with shapes and a key (humans usually) or with pictures and descriptions (plants usually)





Example: Seed colour in peas is controlled by a single gene which has two alleles. **A** represents the dominant allele, and **a** represents the recessive allele.



	A	A
a	Aa	Aa
a	Aa	Aa

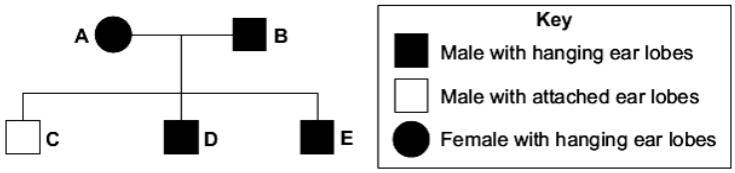
	A	a
a	Aa	aa
a	Aa	aa

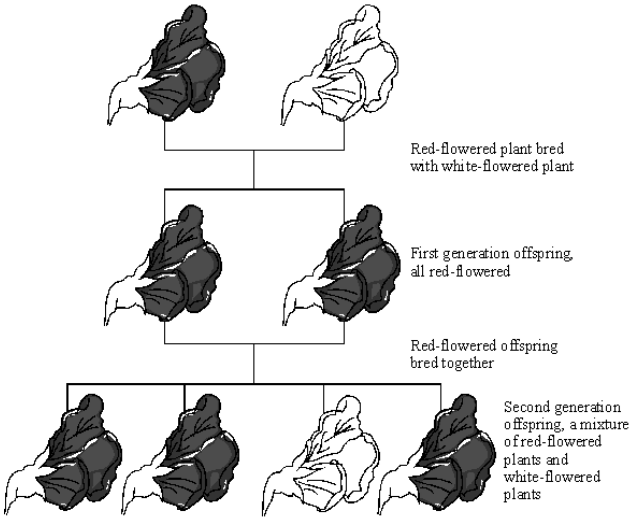
Genetic Diagram Steps:

1. Identify phenotypes – yellow and green
2. Find out which is dominant - yellow
3. Assign allele letters – AA = yellow, aa = green
4. Work out genotypes for first generation – 100% Aa
5. Work out genotypes for second generation – 50% Aa, 50% aa

Part 2: Explain the phenotypes or ratios of offspring using evidence from a genetic diagram or 'family tree'

You can determine if the allele for a characteristic or disorder is dominant or recessive using a family tree diagram. If all children/offspring have the characteristic/disorder, then there is a strong chance the allele for it is dominant.





You can tell if the allele for a characteristic or disorder is recessive if only offspring of heterozygous parents have it e.g. if parents do not have the characteristic/trait but some of their children do. The parents are carriers – you can show this with a punnet square. In both the diagrams above white flowers and attached ear lobes must be caused by recessive alleles whereas red flowers and hanging ear lobes are dominant.

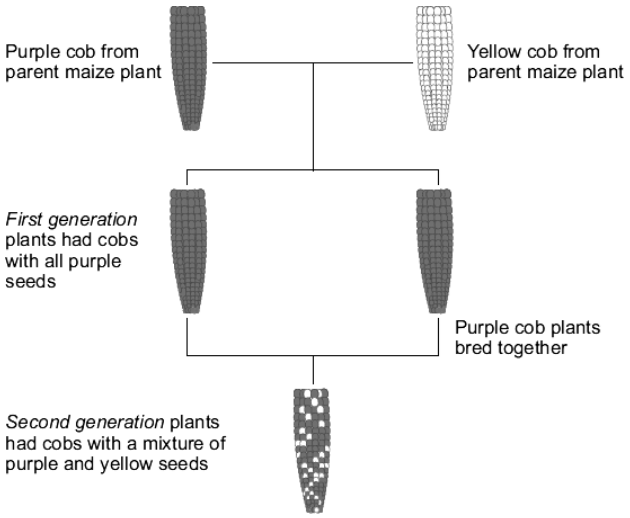
With the ear lobe diagram, there is no evidence from that family tree that females can have attached earlobes so that allele may only be present on the Y chromosome.

With the flower colour diagram you can see that recessive inherited characteristics or traits have the ability to skip generations as there are no white flowers in the second generation of plants.

Worksheet questions: *Answer these in your book, use the information above to help you*

Bronze:

The colour of the seeds is controlled by a gene. The gene has two alleles, purple and yellow. The allele for purple can be represented by the letter **A.** The allele for yellow can be represented by the letter **a**.

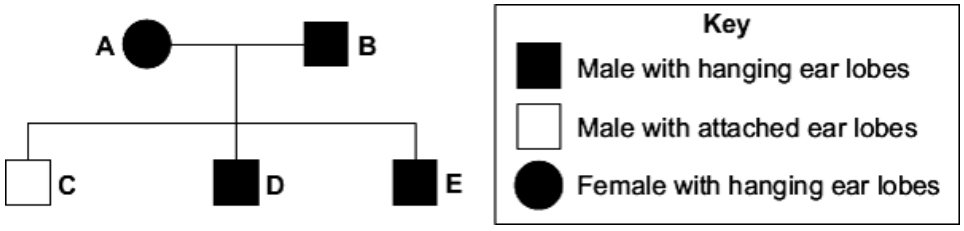


1. All of the first-generation offspring are purple what does this tell you about the purple allele?
2. What alleles does a yellow seed plant have?
   AA Aa aa
3. What will the alleles of the first-generation plants be? Use a Punnett square to help you.
4. The second generation of plants had a mixture of yellow and purple seeds. Can you explain why?

Silver:

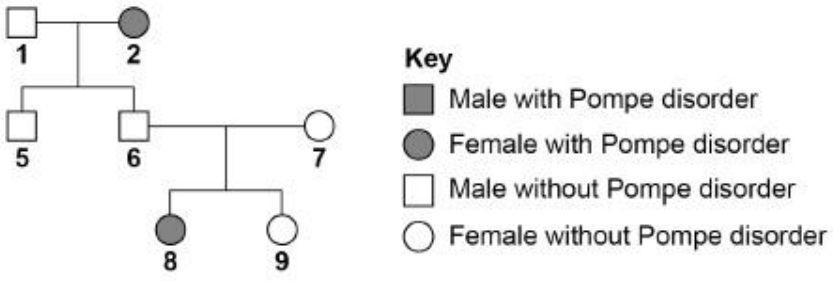
People have different shaped ear lobes, either 'hanging' or 'attached'. A gene controls the shape of a person's ear lobes. Parents A and B both have hanging ear lobes.

1. Draw the symbol for a female with attached ear lobes to complete the key.
2. What does the diagram tell you about the allele for hanging ear lobes? Explain your answer.



Pompe disorder is an inherited condition that affects thousands of people. **R** is the allele for no Pompe disorder and **r** is the allele for Pompe disorder.

1. What is the genotype of person 2?
2. Explain how we can tell Pompe disorder is caused by a recessive allele
3. Person 6 and 7 decide to have another child. What is the probability of 6 and 7 having another child with Pompe disorder?



Gold:

A scientist investigated inheritance in pea plants. The scientist crossed tall pea plants with short pea plants.

T = allele for tall and **t** = allele for short

1. What alleles does plant 1 contain?
2. What will the ratio of tall : short offspring be in the second generation?
3. Two short plants were crossed. This cross produced 100 offspring. How many tall and short plants will there be?