

Inheritance, variation and evolution revision notes

This topic explains how characteristics are passed from parents to offspring, why individuals within a species vary, and how natural selection leads to evolution. It also covers selective breeding and genetic engineering, the evidence for evolution from fossils and resistant bacteria, extinction, and how living things are classified.

For AQA GCSE Combined Science: Trilogy 8464, **Higher tier** is only on the Higher paper.

1 Reproduction Spec 4.6.1

Sexual and asexual reproduction Spec 4.6.1.1

Sexual reproduction involves the joining (fusion) of male and female **gametes**. In animals the gametes are sperm and egg cells. In flowering plants they are pollen and egg cells.

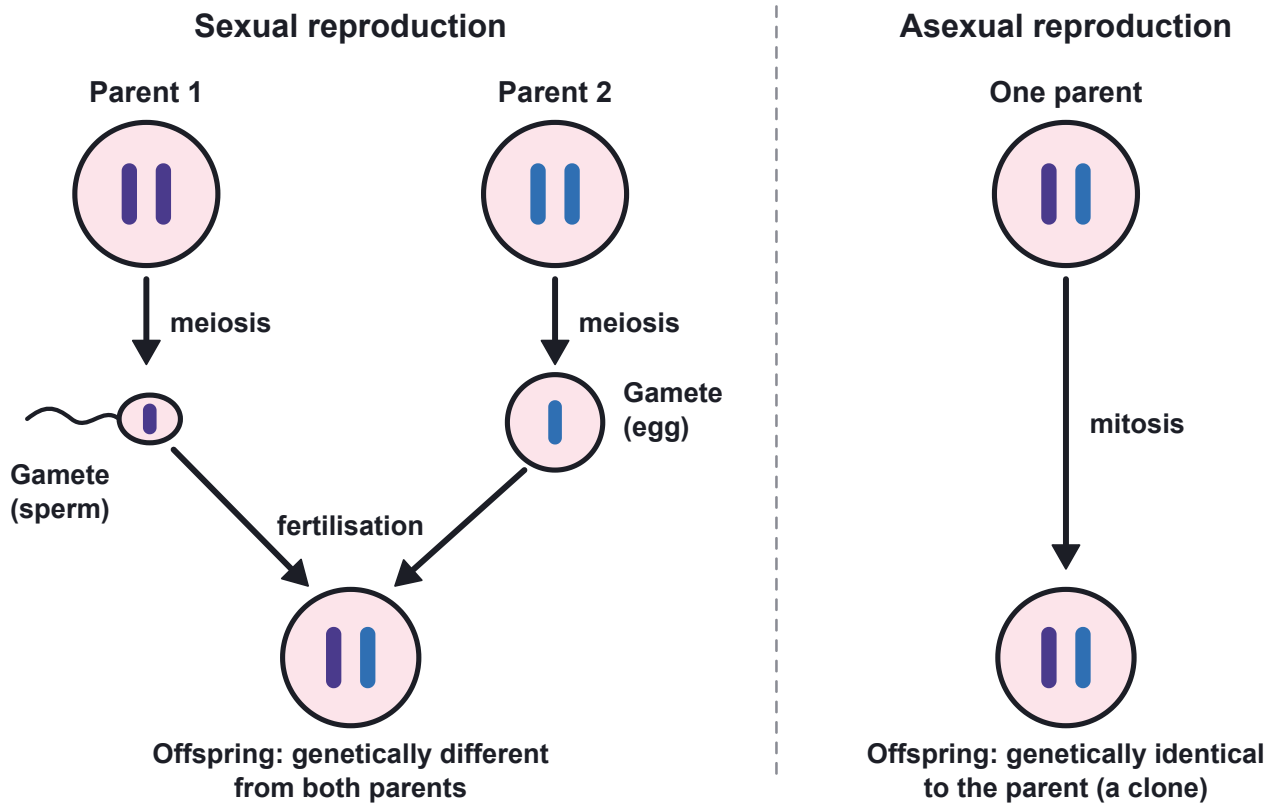
In sexual reproduction there is **mixing of genetic information**, which leads to variety in the offspring. The formation of gametes involves **meiosis**.

Asexual reproduction involves **only one parent** and no fusion of gametes. There is no mixing of genetic information, so the offspring are **genetically identical** to the parent. Genetically identical offspring are called clones. Only mitosis is involved.

Meiosis leads to non-identical cells being formed. Mitosis leads to identical cells being formed.

Sexual and asexual reproduction compared		
	Sexual reproduction	Asexual reproduction
Number of parents	Two	One
Fusion of gametes	Yes	No
Type of cell division used	Meiosis to make gametes	Mitosis only
Mixing of genetic information	Yes	No
Offspring	Genetically different from each other and from the parents	Genetically identical to the parent (clones)

Common mistake: Do not write that asexual offspring are 'similar' to the parent. The mark is for 'genetically identical'.



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Sexual reproduction mixes genetic information from **two parents**; asexual reproduction makes **genetically identical** clones from one parent.

Meiosis Spec 4.6.1.2

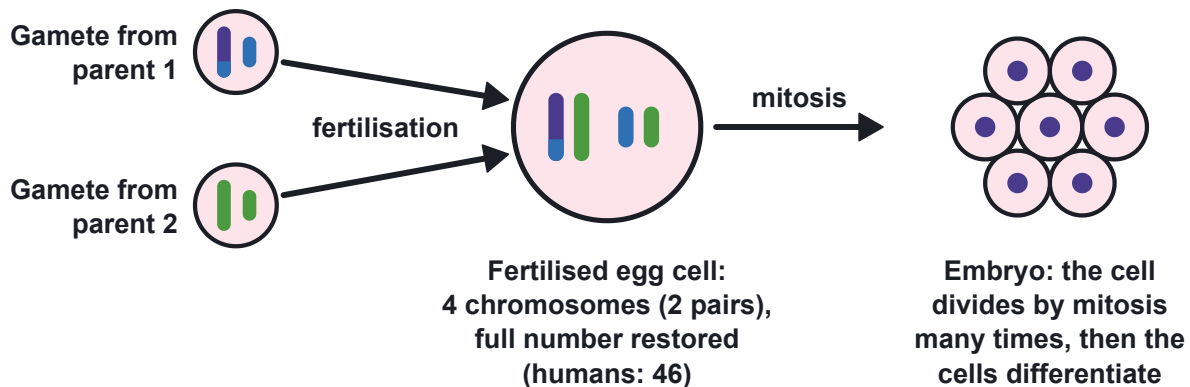
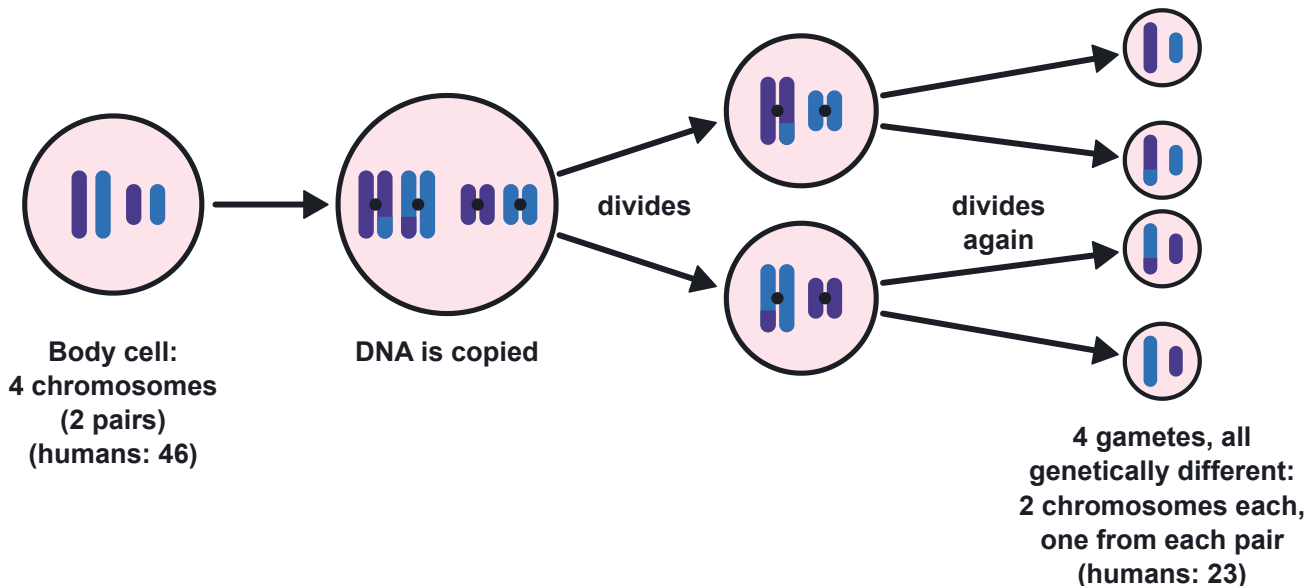
Cells in the **reproductive organs** divide by **meiosis** to form gametes. You need to be able to explain how meiosis halves the number of chromosomes in gametes and how fertilisation restores the full number.

1. The cell copies its genetic information.
2. The cell divides twice to form four gametes.
3. Each gamete has a single set of chromosomes, so the chromosome number is halved.
4. All the gametes are genetically different from each other.

Human body cells contain 23 pairs of chromosomes (46 in total). Each human gamete therefore has 23 chromosomes, one from each pair.

Gametes join at **fertilisation** to restore the normal number of chromosomes. The new cell divides by **mitosis**, so the number of cells increases. As the embryo develops, the cells **differentiate**.

Exam tip: A full explanation has two parts: meiosis halves the chromosome number in the gametes, and fertilisation restores the full number. Knowledge of the stages of meiosis is not required.



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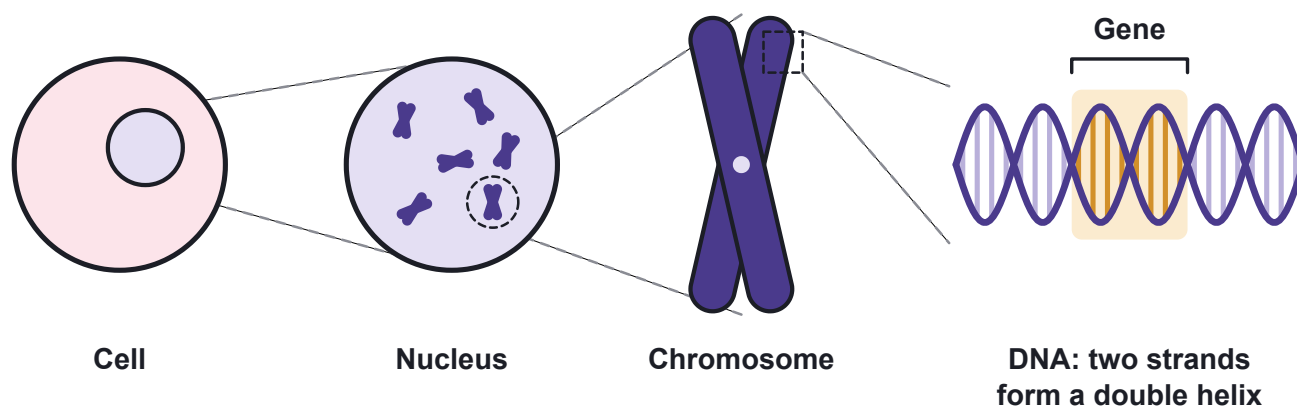
Meiosis **halves** the chromosome number in gametes; fertilisation **restores** the full number, then mitosis builds the embryo.

DNA and the genome Spec 4.6.1.4

The genetic material in the nucleus of a cell is a chemical called **DNA**. DNA is a polymer made up of **two strands forming a double helix**. The DNA is contained in structures called **chromosomes**.

A **gene** is a small section of DNA on a chromosome. Each gene codes for a particular sequence of amino acids, to make a specific protein.

Genome	The entire genetic material of an organism.
Gene	A small section of DNA on a chromosome that codes for a particular sequence of amino acids, to make a specific protein.
Chromosome	A structure in the nucleus that contains DNA.



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A **gene** is a short section of the DNA that makes up a chromosome in the nucleus.

Why the human genome matters

The whole human genome has now been studied, and this will have great importance for medicine in the future. You need to be able to discuss its importance. This is limited to three areas.

- The search for genes linked to different types of disease. Knowing which genes are involved helps scientists understand diseases.
- Understanding and treatment of inherited disorders. Finding the genes involved in a disorder is the first step towards developing treatments.
- Tracing human migration patterns from the past. Comparing the genomes of different groups of people shows how they are related.

Genetic inheritance Spec 4.6.1.6

Gamete	A sex cell (sperm, egg or pollen) that has a single set of chromosomes.
Chromosome	A structure in the nucleus made of DNA, carrying many genes.
Gene	A small section of DNA on a chromosome that codes for a particular protein.
Allele	A different form of the same gene.
Dominant	An allele that is always expressed, even if only one copy is present.
Recessive	An allele that is only expressed if two copies are present (so no dominant allele is present).
Homozygous	Having two alleles that are the same for a trait.
Heterozygous	Having two alleles that are different for a trait.
Genotype	The alleles present for a characteristic.
Phenotype	How the characteristic is expressed, such as black fur.

Some characteristics are controlled by a **single gene**, such as fur colour in mice and red-green colour blindness in humans. Each gene may have different forms called alleles. The alleles present (the

genotype) operate at a molecular level to develop characteristics that are expressed as a phenotype.

Most characteristics are the result of **multiple genes interacting**, rather than a single gene.

Dominant alleles are shown with a capital letter (B) and recessive alleles with the same letter in lower case (b). A black-furred mouse could be BB or Bb. A brown mouse must be bb.

Genetic crosses

A Punnett square shows the possible combinations of alleles from the gametes. Each box is equally likely, so you can use it to predict **probability** and ratios. Probability tells you the chance of an outcome, not what will actually happen in a small family.

Cross between two heterozygous mice

In mice, black fur (B) is dominant to brown fur (b). Two black mice, both Bb, are crossed. Show the possible offspring, the ratio of phenotypes and the probability of a brown mouse.

1. Gametes from one parent: B and b. Gametes from the other parent: B and b.
2. Fill in the Punnett square: BB, Bb, Bb, bb.
3. Three of the four boxes contain at least one B, so they are black. One box is bb, so it is brown.
4. Ratio of black : brown = 3 : 1.
5. Probability of brown = 1 out of 4.

Answer: Ratio 3 black : 1 brown. The probability of a brown mouse is 1 in 4, which is 0.25 or 25%.

Using a ratio

The cross above produces 80 offspring in total. About how many would you expect to be black?

1. The ratio is 3 : 1, so there are $3 + 1 = 4$ parts.
2. One part = $80 \div 4 = 20$.
3. Black = 3 parts = $3 \times 20 = 60$.

Answer: About 60 black offspring.

Maths skill: Use direct proportion and simple ratios to express the outcome of a cross. A 3 : 1 ratio means a probability of $\frac{3}{4}$ for the dominant phenotype and $\frac{1}{4}$ for the recessive. Real results are rarely exact.

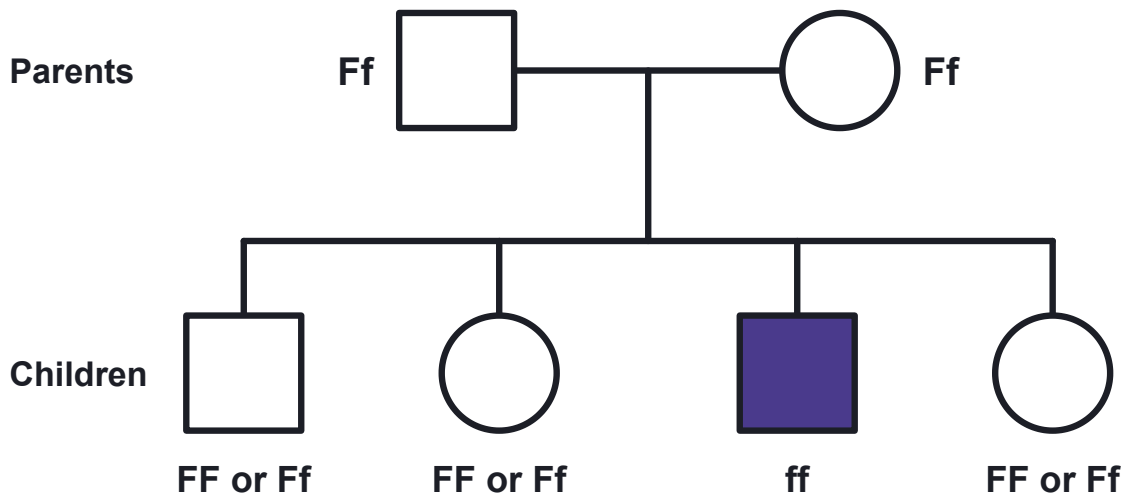
You must be able to complete a Punnett square diagram and extract and interpret information from genetic crosses.

Higher tier

Higher tier students must also be able to **construct** a genetic cross by Punnett square diagram from scratch and use it to make predictions using the theory of probability.

Family trees

A family tree shows how a characteristic is passed through a family. Squares usually show males and circles show females, and shading shows people who have the characteristic. If two parents who do not show a characteristic have a child who does, the characteristic must be **recessive**, and both parents must be heterozygous.



Unaffected male



Unaffected female



Affected male



Affected female

F = dominant allele

f = recessive allele

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Two unaffected parents have an affected child, so the allele is **recessive** and both parents are **Ff**.

Exam tip: In a Punnett square, write the alleles you are using in a key (for example B = black, b = brown), put the gametes on the outside and the offspring genotypes inside the boxes.

Inherited disorders Spec 4.6.1.7

Some disorders are inherited. They are caused by the inheritance of certain alleles.

- **Polydactyly** (having extra fingers or toes) is caused by a **dominant** allele.
- **Cystic fibrosis** (a disorder of cell membranes) is caused by a **recessive** allele.

Polydactyly

Polydactyly is caused by a dominant allele (D). One parent has polydactyly and is Dd. The other parent does not have polydactyly. What is the chance that a child has polydactyly?

1. A person without polydactyly must be dd (a dominant allele is always expressed).
2. Gametes: D and d from one parent, d and d from the other.
3. Offspring: Dd, Dd, dd, dd.
4. Two of the four have a D allele, so two of the four have polydactyly.

Answer: The ratio of polydactyly : no polydactyly is 1 : 1, so the chance of a child having polydactyly is 1 in 2 (50%).

Cystic fibrosis

Cystic fibrosis is caused by a recessive allele (f). Two parents do not have cystic fibrosis but both are carriers (Ff). What is the chance that a child has cystic fibrosis?

1. Gametes from each parent: F and f.
2. Offspring: FF, Ff, Ff, ff.
3. Only ff has two recessive alleles, so only ff has cystic fibrosis.

Answer: 1 in 4 (25%). The ratio of unaffected to affected is 3 : 1.

Embryo screening and gene therapy

Embryos can be **screened** for faulty alleles before they are implanted, so that only embryos without the disorder are used. Modern medical research is also exploring gene therapy, which is genetic modification to overcome some inherited disorders. Both may alleviate suffering, but they raise ethical issues.

You may be asked to make an informed judgement about the economic, social and ethical issues, using the information given.

Embryo screening: points to weigh up

	For	Against
Ethical	Prevents suffering for the child and family	Embryos that would have the disorder are destroyed, which some people believe is wrong
Social	Fewer children born with serious disorders	May lead to pressure to choose other characteristics; people living with the disorder may feel devalued
Economic	May reduce the long-term cost of treatment and care	The screening itself is expensive and is not available to everyone

Exam tip: In 'evaluate' or 'discuss' questions, give points for both sides and finish with a judgement that uses the information given in the question.

Sex determination Spec 4.6.1.8

Ordinary human body cells contain **23 pairs of chromosomes**. **22 pairs control characteristics only**, but one of the pairs carries the genes that determine sex.

- In females the sex chromosomes are the same: **XX**.
- In males the sex chromosomes are different: **XY**.

All eggs contain an X chromosome. Half of the sperm contain an X and half contain a Y. The sperm that fertilises the egg decides the sex of the baby.

Sex inheritance cross

Show the cross between a female (XX) and a male (XY) and state the ratio of female to male offspring.

1. Gametes from the mother: X and X. Gametes from the father: X and Y.
2. Punnett square: XX, XX, XY, XY.
3. Two of the four boxes are XX (female) and two are XY (male).

Answer: Ratio of female : male = 1 : 1. The probability of each child being a girl, or a boy, is 50%.

Common mistake: Each fertilisation is independent. If a couple already have three girls, the chance of a boy next time is still 50%.

Quick check

1. Why are the offspring of asexual reproduction genetically identical to the parent?
2. How does meiosis halve the number of chromosomes?
3. Define the genome of an organism.
4. What is the difference between genotype and phenotype?
5. What are the sex chromosomes of a human female and male?

2 Variation and evolution Spec 4.6.2

Variation Spec 4.6.2.1

Differences in the characteristics of individuals in a population are called **variation**. Variation may be due to differences in:

- the genes they have inherited (genetic causes)
- the conditions in which they have developed (environmental causes)
- a combination of genes and the environment.

The phenotype of an organism develops from its genome and the way the genome **interacts with the environment**.

Causes of variation	
Cause	Examples
Genetic	Eye colour, blood group
Environmental	A scar from an injury; a plant that grows poorly in poor soil
Both	Body mass or height: genes set a potential, but diet and lifestyle affect what is reached

Mutations

There is usually extensive genetic variation within a population of a species. All variants arise from **mutations**. Of these:

- most have **no effect** on the phenotype
- some **influence** the phenotype
- very few **determine** the phenotype.

Mutations occur continuously. Very rarely a mutation leads to a new phenotype. If the new phenotype is suited to an environmental change, it can lead to a relatively rapid change in the species.

Exam tip: 'Mutation' is the only source of new variants. Sexual reproduction shuffles existing alleles into new combinations, but mutations are where new alleles come from.

Evolution Spec 4.6.2.2

Evolution	A change in the inherited characteristics of a population over time through a process of natural selection, which may result in the formation of a new species.
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The theory of evolution by natural selection states that all species of living things have evolved from simple life forms that first developed more than **three billion years ago**.

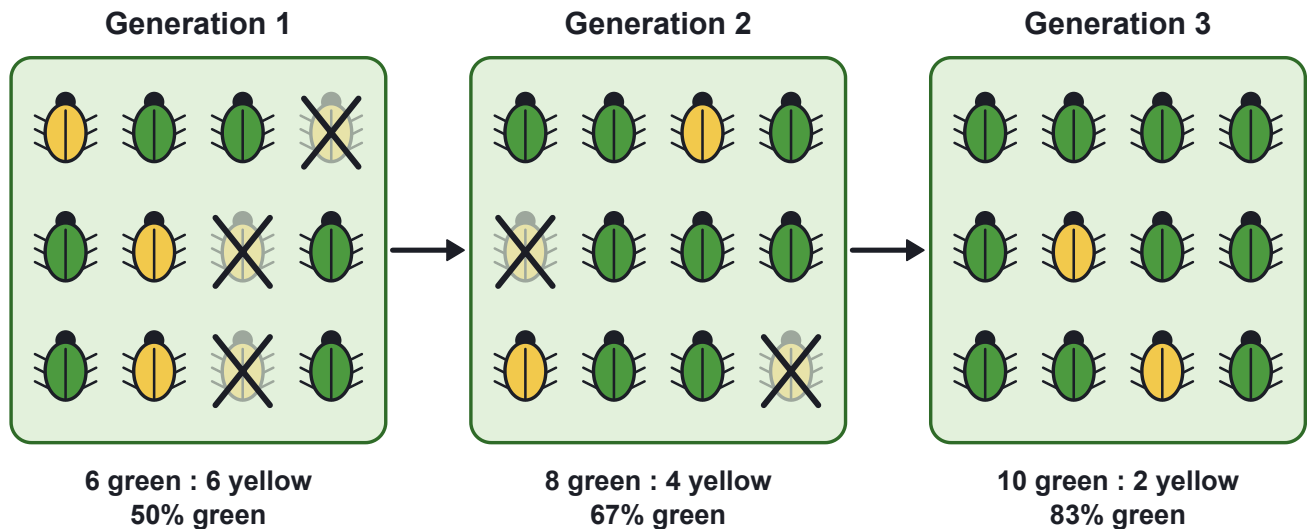
How natural selection works

1. Individuals in a population show variation in their phenotypes, caused by differences in their genes.
2. The environment puts pressure on the population. For example, there may be predators, disease or competition for food.
3. Individuals with phenotypes **best suited** to their environment are more likely to **survive and breed**.
4. They pass on the alleles for the useful characteristic to their offspring.
5. Over many generations, a greater proportion of the population has the characteristic.

New species

If two populations of one species become so different in phenotype that they can no longer **interbreed to produce fertile offspring**, they have formed two new species.

Common mistake: Individuals do not 'adapt' or 'evolve' during their lives. Populations evolve, because the better-suited variants survive and reproduce. Do not write that organisms 'need' a characteristic so they develop it.



= eaten by birds. Yellow beetles are easier to see on green leaves, so they are more likely to be eaten. Green beetles are more likely to survive, breed and pass on their alleles, so the proportion of green beetles rises.

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Better-suited individuals are more likely to **survive and breed**, so their alleles become more common in each generation.

Selective breeding Spec 4.6.2.3

Selective breeding (artificial selection) is the process by which humans breed plants and animals for particular genetic characteristics. Humans have been doing this for thousands of years, since they first bred food crops from wild plants and domesticated animals.

1. Choose parents with the **desired characteristic** from a mixed population.
2. Breed them together.
3. From the offspring, choose those with the desired characteristic and breed them together.
4. Continue over **many generations** until all the offspring show the desired characteristic.

The characteristic can be chosen for usefulness or appearance. Examples:

- disease resistance in food crops
- animals which produce more meat or milk
- domestic dogs with a gentle nature
- large or unusual flowers.

Impact of selective breeding

Benefits and risks	
Benefits	Risks
Crops and animals that give higher yields of food	Can lead to inbreeding , where some breeds are particularly prone to disease or inherited defects
Disease-resistant crops	Reduces the number of different alleles in the population, because only a few individuals are bred
Animals or plants with useful or attractive features	Some people have ethical objections to animals being bred for human use

Exam tip: Always say what is selected and what happens next: 'select the cows that give most milk, breed them, select the best of the offspring, repeat over many generations'.

Genetic engineering Spec 4.6.2.4

Genetic engineering	A process which involves modifying the genome of an organism by introducing a gene from another organism to give a desired characteristic.
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In genetic engineering, genes from the chromosomes of humans and other organisms can be 'cut out' and transferred to cells of other organisms.

- Plant crops have been genetically engineered to be resistant to diseases or to produce bigger, better fruits.
- Bacterial cells have been genetically engineered to produce useful substances such as **human insulin** to treat diabetes.

Crops that have had their genes modified in this way are called **genetically modified (GM) crops**. GM crops include ones that are resistant to insect attack or to herbicides. GM crops generally show increased yields.

Benefits, risks and objections

Genetic engineering in agriculture and medicine		
	Potential benefits	Potential risks and objections
Agriculture	Increased yields; resistance to insects, herbicides or disease; bigger, better fruits	Effects on populations of wild flowers and insects; some people feel the effects of eating GM crops on human health have not been fully explored
Medicine	Bacteria can make useful substances such as human insulin; research is exploring genetic modification to overcome some inherited disorders	Some people object to altering the genes of organisms, especially humans; long-term effects may not be known

Some people have objections to genetic engineering, even though it has huge potential benefits.

Higher tier

The main steps in genetic engineering

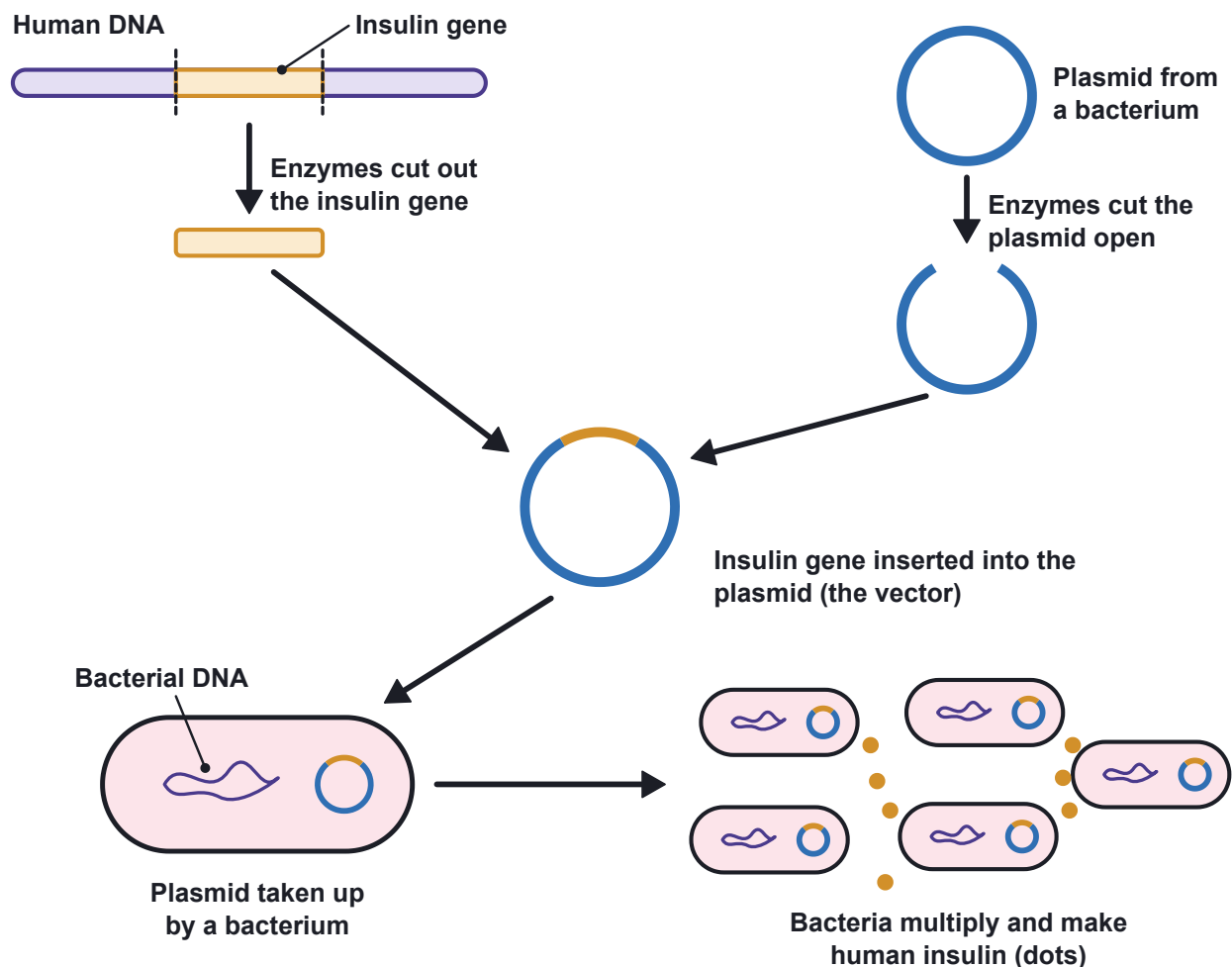
Higher tier

1. **Enzymes** are used to isolate the required gene.
2. The gene is inserted into a **vector**, usually a bacterial plasmid or a virus.
3. The vector is used to insert the gene into the required cells.
4. Genes are transferred to the cells of animals, plants or microorganisms at an **early stage in their development**, so that they develop with the desired characteristics.

Higher tier

Exam tip: Use the key words in order: enzymes isolate the gene, the gene goes into a vector (plasmid or virus), the vector puts it into the cells, at an early stage of development.

Higher tier



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Enzymes isolate the gene, a **plasmid (vector)** carries it into a bacterium, and the bacteria multiply and make **human insulin**.

Quick check

- 1 . Give the three possible causes of variation.
- 2 . What do all variants arise from?
- 3 . When have two populations become two new species?
- 4 . Name one problem caused by selective breeding.
- 5 . What is a GM crop?

3 The development of understanding of genetics and evolution

Spec 4.6.3

Evidence for evolution Spec 4.6.3.4

The theory of evolution by natural selection is now **widely accepted**. Evidence for Darwin's theory is now available. You need to be able to describe the evidence, including fossils and antibiotic resistance in bacteria.

- It has been shown that **characteristics are passed on to offspring in genes**.
- The **fossil record** provides further evidence (see the next section).
- Knowledge of how **resistance to antibiotics evolves in bacteria** provides evidence that natural selection happens and can be seen in a short time (see Resistant bacteria).

Exam tip: Link your evidence to the theory. Say what is seen (for example fossils show how organisms have changed), then say how it supports evolution by natural selection.

Fossils Spec 4.6.3.5

Fossils are the 'remains' of organisms from millions of years ago, which are found in rocks.

How fossils may form

- From parts of organisms that have **not decayed** because one or more of the conditions needed for decay are absent.
- When parts of the organism are **replaced by minerals** as they decay.
- As **preserved traces** of organisms, such as footprints, burrows and rootlet traces.

We can learn from fossils how much or how little different organisms have changed as life developed on Earth.

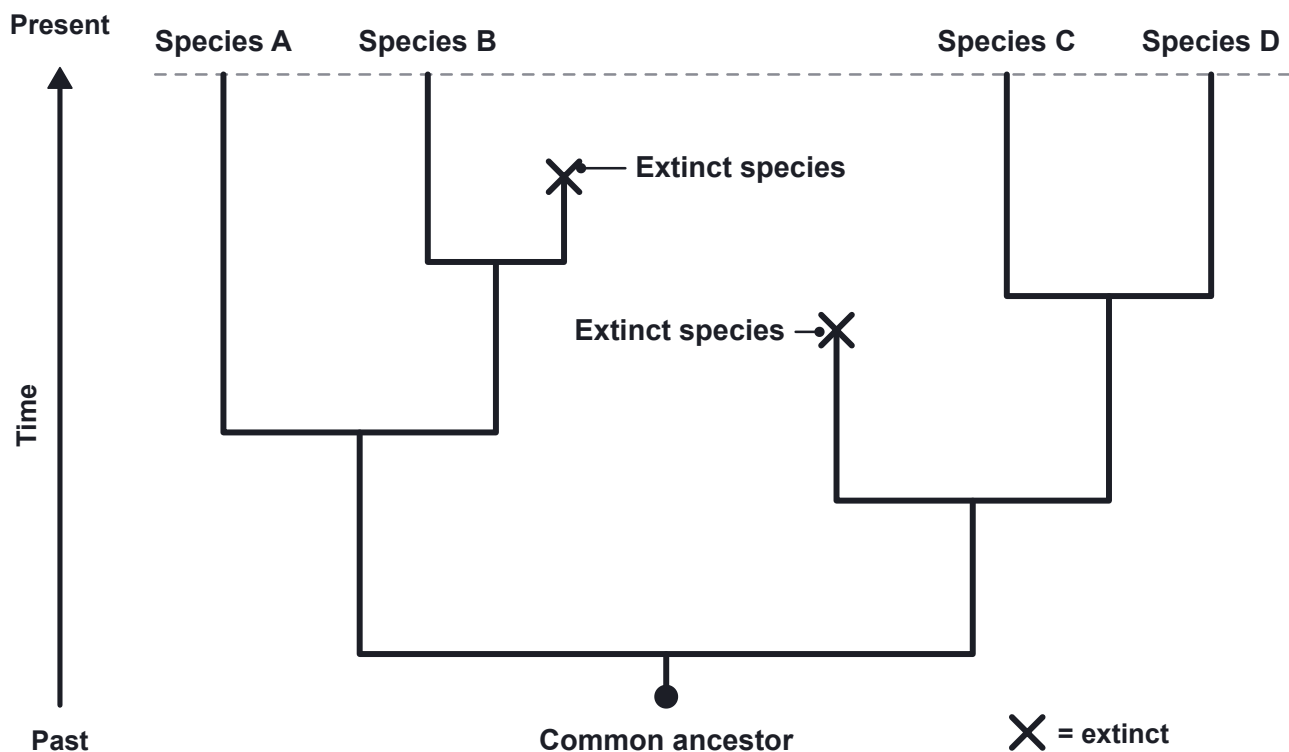
Why the fossil record is incomplete

Many early forms of life were **soft-bodied**, which means that they have left few traces behind. What traces there were have been mainly **destroyed by geological activity**. This is why scientists cannot be certain about how life began on Earth.

Evolutionary trees

You may need to extract and interpret information from charts, graphs and tables, such as evolutionary trees. Evolutionary trees use fossil data for extinct organisms and current classification data for living organisms.

Exam tip: Fossils are evidence for how organisms have changed over time, but gaps mean the record is incomplete. Quote both ideas in an evaluation.



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Branches ending in a cross are **extinct** species, known only from fossils.

Extinction Spec 4.6.3.6

Extinction occurs when there are no remaining individuals of a species still alive.

You need to be able to describe factors that may contribute to the extinction of a species. A species may become extinct because of:

- a change to the environment over geological time
- new **predators**
- new **diseases**
- new, more successful **competitors**
- a single catastrophic event, such as a massive volcanic eruption or an asteroid collision.

Exam tip: Always link the factor to the species: for example, a new disease kills individuals faster than they can reproduce, so the numbers fall until none are left.

Resistant bacteria Spec 4.6.3.7

Bacteria can evolve rapidly because they **reproduce at a fast rate**. Mutations of bacterial pathogens produce new strains.

1. A mutation produces a strain that is resistant to antibiotics.
2. When antibiotics are used, non-resistant bacteria are killed. The resistant bacteria are not killed.
3. The resistant bacteria **survive and reproduce**, so the population of the resistant strain rises.
4. The resistant strain spreads, because people are not immune to it and there is no effective treatment.

MRSA is resistant to antibiotics.

Reducing the rate of development of resistant strains

- Doctors should not prescribe antibiotics inappropriately, such as for non-serious or viral infections.
- Patients should complete their course of antibiotics so all bacteria are killed and none survive to mutate and form resistant strains.
- The agricultural use of antibiotics should be restricted.

The development of new antibiotics is costly and slow. It is unlikely to keep up with the emergence of new resistant strains.

Common mistake: People do not become resistant to antibiotics. The bacteria are resistant. The resistant bacteria survive and reproduce, passing on the resistance.

Quick check

1. Why is the fossil record incomplete?
2. Why do resistant strains of bacteria spread so quickly?
3. Give one way to reduce the development of antibiotic resistance.

4 Classification of living organisms Spec 4.6.4

The Linnaean system and binomial names Spec 4.6.4

Traditionally, living things have been classified into groups depending on their structure and characteristics, in a system developed by **Carl Linnaeus**.

Linnaeus classified living things into: **kingdom, phylum, class, order, family, genus and species**. Organisms are named by the **binomial system** of genus and species.

Example: the binomial name of humans	
Level	Name
Genus	<i>Homo</i>
Species	<i>sapiens</i>
Binomial name	<i>Homo sapiens</i>

In a binomial name, the genus is written first with a capital letter, and the species second with a lower-case letter. Binomial names are written in italics or underlined.

Exam tip: A memory aid for the order: King Philip Came Over For Good Soup. In questions you are given information, so you need to show you understand how the system works.

Developments in classification Spec 4.6.4

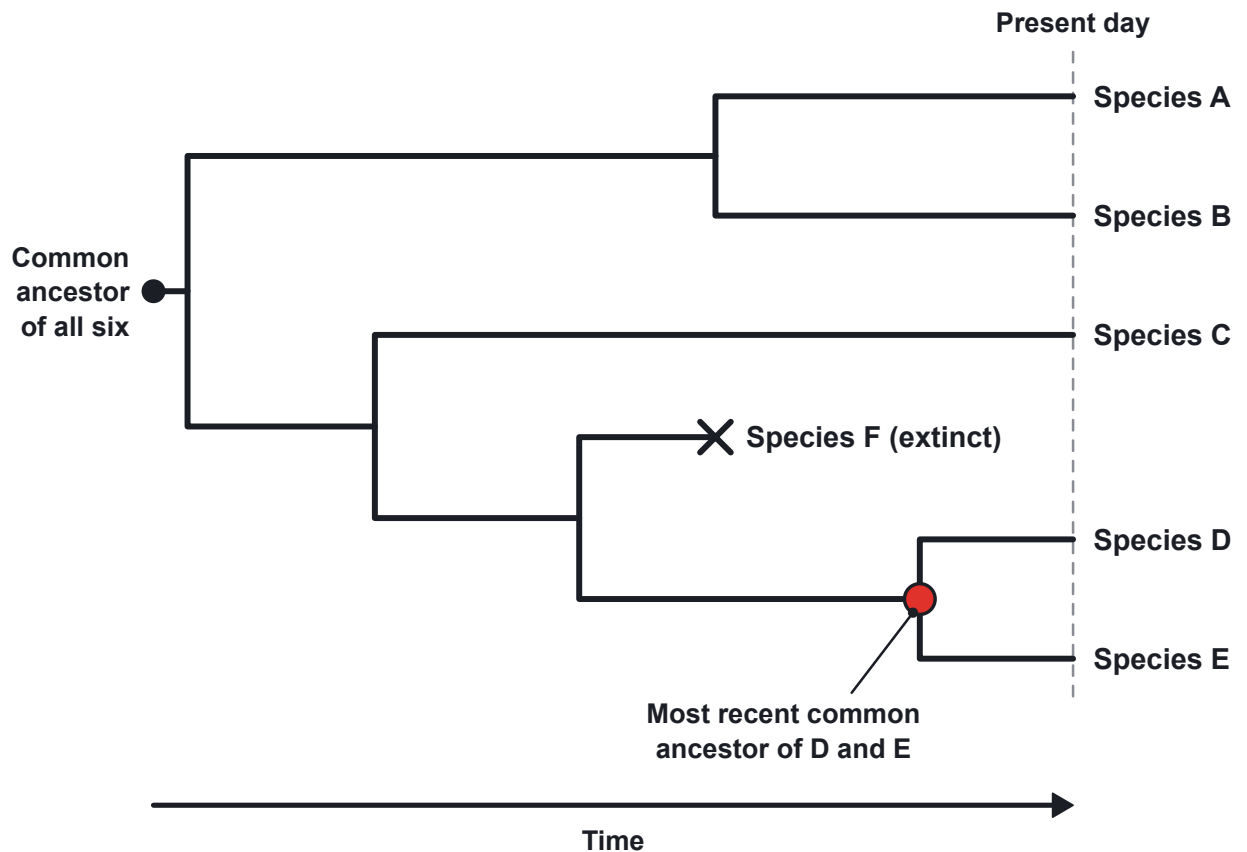
As evidence of internal structures became more developed due to improvements in **microscopes**, and the understanding of **biochemical processes** progressed, new models of classification were proposed.

Due to evidence available from **chemical analysis**, there is now a **three-domain system** developed by Carl Woese. In this system, organisms are divided into:

The three-domain system	
Domain	Description
Archaea	Primitive bacteria, usually living in extreme environments
Bacteria	True bacteria
Eukaryota	Includes protists, fungi, plants and animals

Evolutionary trees

Evolutionary trees are a method used by scientists to show how they believe organisms are related. They use current classification data for living organisms and fossil data for extinct organisms. The more recently two branches split from a common ancestor, the more closely related the organisms are.



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D and E share the **most recent common ancestor**, so they are the most closely related species on this tree.

Exam tip: To read a tree, find the point where two branches split. The nearer that point is to the present, the more closely related the two species are.

Quick check

1. List the Linnaean groups from largest to smallest.
2. Which two groups make up a binomial name?
3. Name the three domains in Woese's system.
4. What data are used to build evolutionary trees?

Quick check answers

Reproduction

1. There is only one parent and no fusion of gametes, and only mitosis is involved.
2. The cell divides twice to form four gametes, each with a single set of chromosomes.
3. The entire genetic material of that organism.
4. Genotype is the alleles present; phenotype is how the characteristic is expressed.
5. Female XX, male XY.

Variation and evolution

1. Genetic causes, environmental causes, or a combination of genes and the environment.
2. Mutations.
3. When they are so different in phenotype that they can no longer interbreed to produce fertile offspring.
4. Inbreeding, where some breeds are prone to disease or inherited defects.
5. A crop that has had its genes modified by genetic engineering.

The development of understanding of genetics and evolution

1. Early life was soft-bodied and left few traces, and many traces were destroyed by geological activity.
2. Bacteria reproduce quickly, people are not immune and there is no effective treatment.
3. Do not prescribe antibiotics inappropriately, complete the course, or restrict agricultural use.

Classification of living organisms

1. Kingdom, phylum, class, order, family, genus, species.
2. Genus and species.
3. Archaea, bacteria and eukaryota.
4. Current classification data for living organisms and fossil data for extinct organisms.