

Reproduction

Revision notes • Sexual and asexual reproduction, meiosis, DNA and the genome, genetic crosses, inherited disorders and sex determination.

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

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.

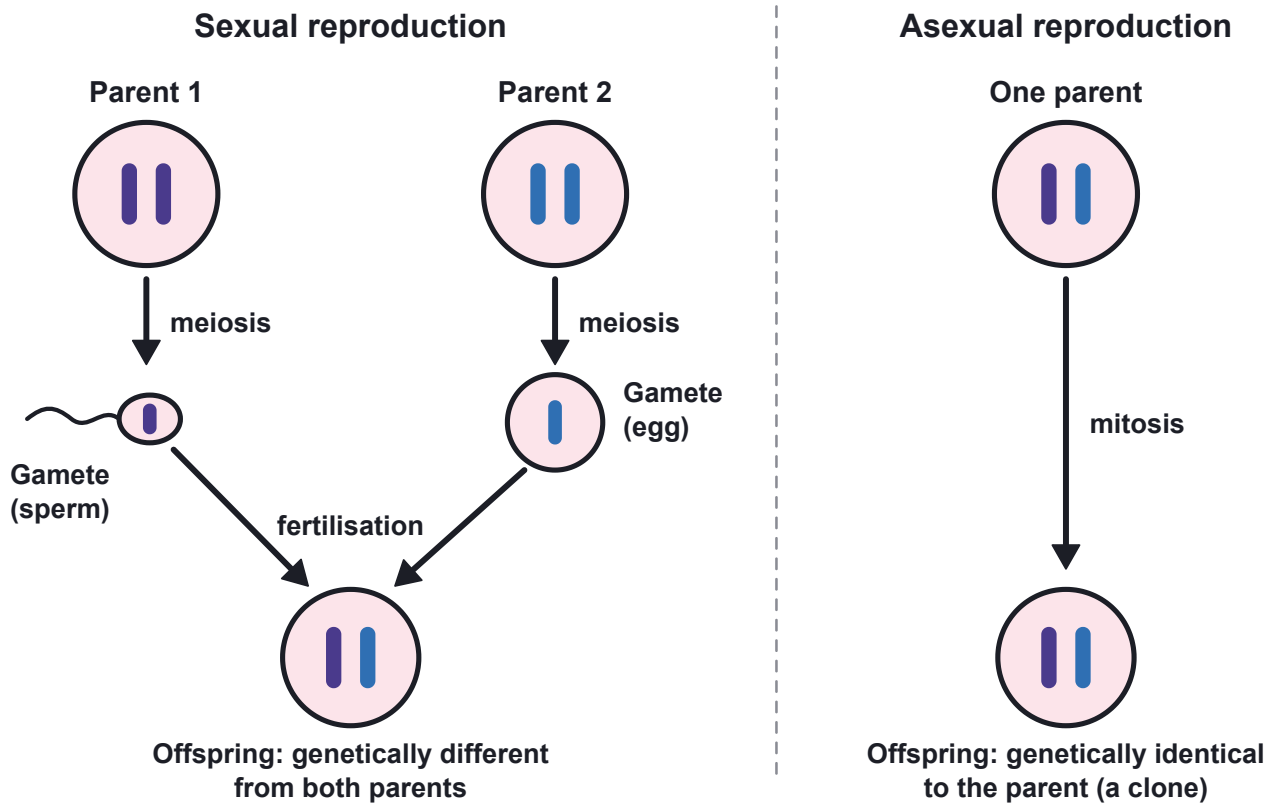
In sexual reproduction there is **mixing of genetic information**, which leads to variety in the offspring. The formation of gametes involves **meiosis**.

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

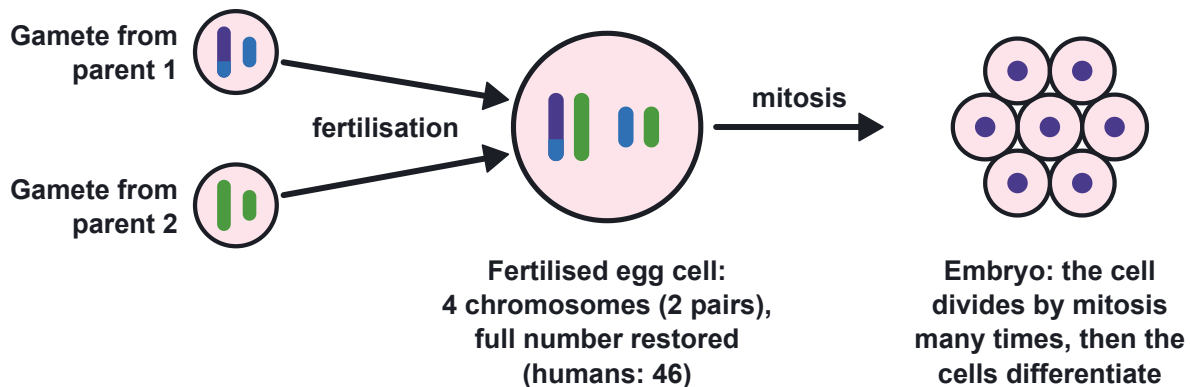
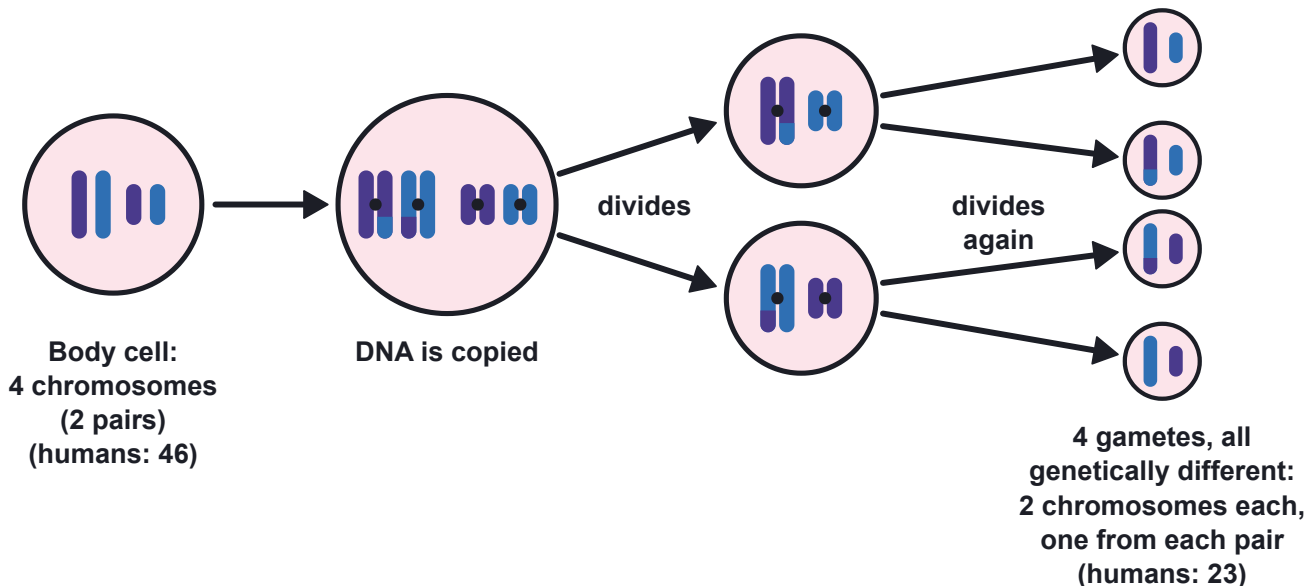
Cells in the **reproductive organs** divide by **meiosis** to form gametes. You need to be able to explain how meiosis halves the number of chromosomes in gametes and how fertilisation restores the full number.

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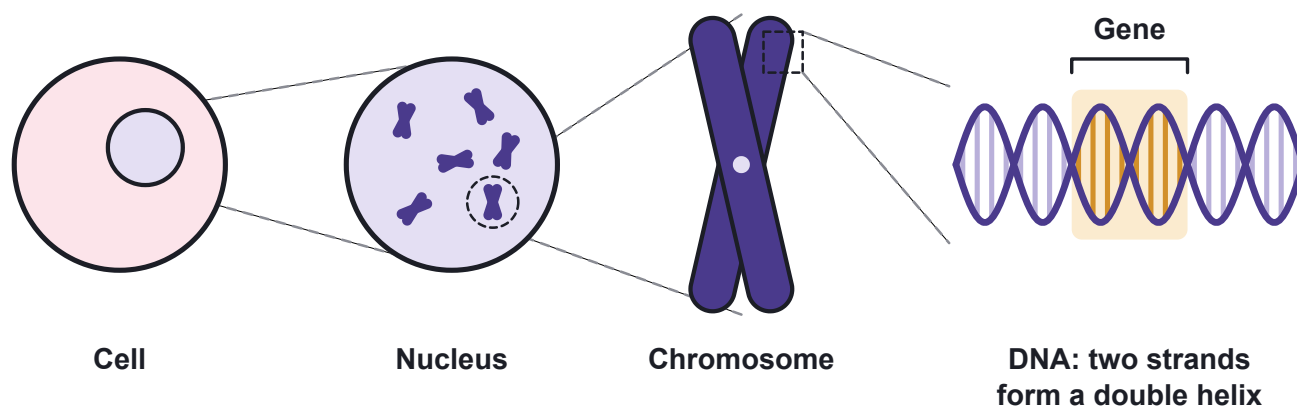
Meiosis **halves** the chromosome number in gametes; fertilisation **restores** the full number, then mitosis builds the embryo.

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.

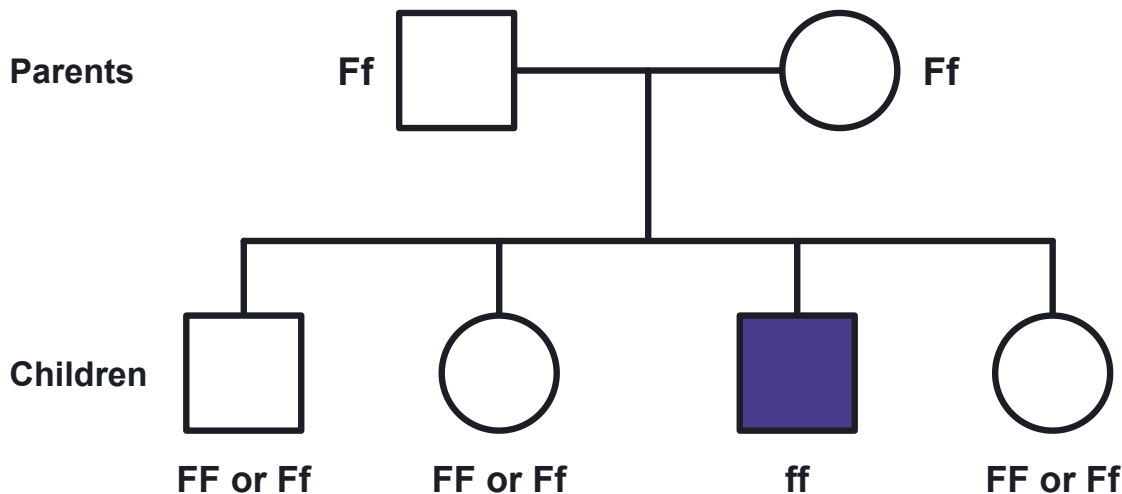
You must be able to complete a Punnett square diagram and extract and interpret information from genetic crosses.

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?

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.