# Biology Knowledge Organiser
# B13 - Reproduction

## Genetic inheritance

All the genes you have, you inherited from your parents. They gave you half your genome each. Since they gave you one from each pair of chromosomes you have now, they in fact gave you one copy of each gene each – i.e. genes for the same thing. We call the two different versions of each gene **alleles**. Some characteristics are controlled by one gene – or rather, the two alleles of a single gene. E.g. fur colour in mice, red-green colour blindness in humans. However, most characteristics come about thanks to many genes and their interactions, not just one gene.

The alleles present in an individual organism cause body cells to produce certain proteins, or versions of proteins (as this is what a gene does remember). This is called **expression** of a gene, and leads to physical characteristics we call **phenotypes**.

This is easier with an example. Look at the cats below: the allele for short fur in cats is dominant to the allele for long fur. Let's call the alleles F and f respectively. In the top example, both parents are homozygous dominant (genotype: FF). This means all the gametes they produce will have one F in them, so at fertilisation the only possibility is for the offspring to get FF. So all their offspring have the short fur **phenotype**.

In the second row, both parents have long hair, so they must both have the genotype ff (homozygous recessive). Consequently, all their offspring must have long hair too.

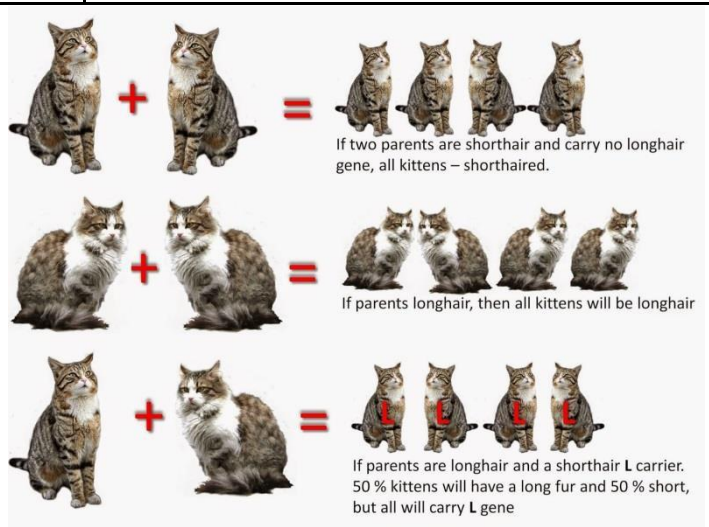
In the third row, the first parent has short hair but is heterozygous (genotype: Ff) – so they still have short hair as the short hair allele is dominant. If they mate with a long hair cat (genotype ff), there are different probabilities for offspring phenotypes, as they will get *either* F or f from the first parent. So, half of them will have short hair (with genotype Ff) and half will have long hair (with genotype ff).

## Probability and ratios

Knowing the genotypes of the parents allows you to work out the **probability** of each genotype (and therefore phenotype) in the offspring. It does not guarantee, like in the bottom cat example, that they'll have four kittens, or that half will have long hair. What it tells us is: for each kitten, there is a 50% chance of it having long hair.

The other way of saying this is that the **expected ratio** of offspring genotypes is 1:1 for long:short hair. So if the bottom two cat parents had 50 kittens, we'd expect 25 of each hair length.

| Key Terms | Definitions |
|---|---|
| Allele | A form or version of a gene. Since you inherit a copy of each chromosome from each parent, you have two copies of each gene – we call these two versions alleles. |
| Express | In genetics, to 'express' a gene means for it to be used by the body to make a protein, causing a characteristic. |
| Dominant | Describes alleles that are always expressed (so you see the effects in the organism). Indicated with a capital letter to represent the allele e.g. D. |
| Recessive | Describes alleles that are only expressed if there are two recessive copies (one from each parent). In other words, recessive alleles are only expressed if there is **no** dominant allele present. Indicated with a lower case letter to represent the allele e.g. d. |
| Genotype | The combination of alleles that an individual has. Often represented with two letters: e.g. DD, Dd or dd. |
| Phenotype | The physical characteristic that results from a particular genotype. |
| Homozygous | Describes a genotype where both alleles are the same – e.g. DD is homozygous dominant; dd is homozygous recessive. |
| Heterozygous | Describes a genotype where the two alleles are different (one dominant, one recessive) – e.g. Dd. |



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## Inherited disorders

Some disorders (or diseases – same thing really) are inherited, so we can also call them **genetic disorders**. If someone inherits a certain allele/combination of alleles, they have the inherited disorder. Two examples to know:

- **Polydactyly**: a condition where people have extra fingers or toes. This is caused by a dominant allele, so only one copy is needed to have the condition.
- **Cystic fibrosis**: a condition where protein pumps in cell membranes don't work properly, leading to thick and sticky mucus being produced in the lungs and intestines. This is caused by a recessive allele, so individuals with cystic fibrosis are all homozygous recessive.

Studying family trees can help genetic scientists decide whether a disorder is caused by a recessive or dominant allele. In the family tree shown, C is the allele for healthy cell membranes, and c is the allele for disordered cell membranes. Both parents must have at least one c to have children with cystic fibrosis, as the family tree shows. (Note: anyone without a genotype shown has the genotype CC).

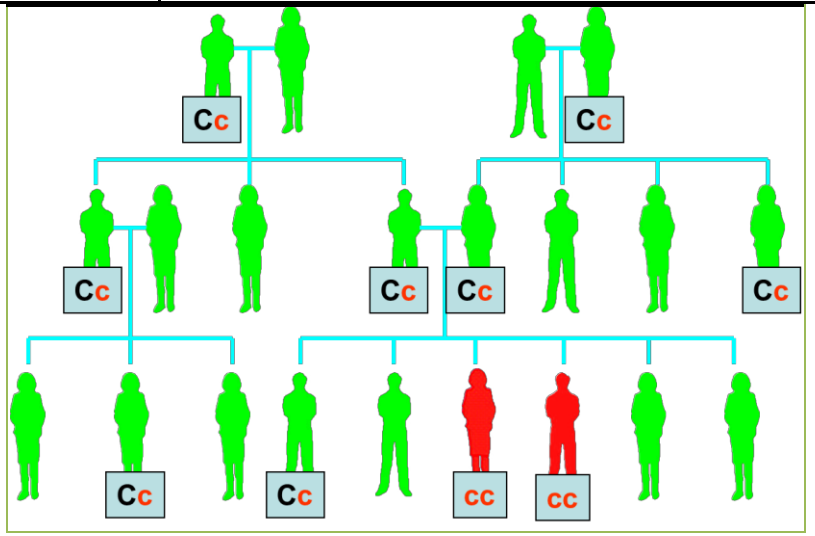
Since we know which alleles cause conditions like these, unborn babies, or embryos produced during IVF, can be checked – or **screened** – to see if they have the inherited disorder. This practice, *embryo screening*, can be used to inform whether an embryo should be implanted in IVF, or, if used during pregnancy, to decide whether an abortion should take place. Obviously, these are huge decisions and the right to life of the embryo must be weighed against the difficulties they'll face with an inherited condition and the personal choice and beliefs of the parents.

## Sex determination

In biology, sex is not short for sexual intercourse. Sex means male or female – so is only relevant to organisms that reproduce sexually. The sex of offspring is determined by the combination of sex chromosomes inherited from the parents. Of the 23 pairs of chromosomes all humans have, 22 control body characteristics and the 23rd pair determines sex. [Note: like all chromosomes, the sex chromosomes carry genes, they just have the extra function of sex determination.] Human females have the combination for pair 23: XX. We say they have two X chromosomes. Human males have the combination XY for pair 23 (they are different).

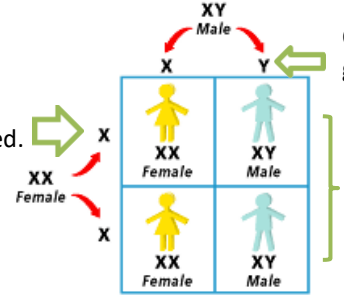
When having children, then, mothers always pass on one X chromosome to their offspring. Males can pass on an X chromosome OR a Y chromosome – there's a 50:50 chance of each. This is because, when cells divide by meiosis to make gametes, all the female gametes contain an X, but half the sperm cells have an X, half have a Y. How these combine to give a 50% chance of a girl is shown in the **Punnett square** to the right.

| Key Terms | Definitions |
|---|---|
| Screening | The practice of checking for a disease or an inherited disorder. |
| Carrier | An individual with one copy of the recessive allele that causes an inherited disorder (e.g. Cc for the cystic fibrosis genotype). As a result, they don't have the disorder, but they can pass one allele for it onto their offspring. |
| Sex chromosomes | Pair 23 in humans. Females have the combination XX, males have the combination XY. |
| Genetic cross | An unglamorous term given to mating between two individuals, producing offspring. |
| Punnett square | A tool used to predict the outcome of a genetic cross. |




On the side, the possible female gametes are placed.

On the top, the possible male gametes are placed.




The possible combinations are shown. The ratio of female:male is 2:2, simplifying to 1:1.

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## Types of reproduction

Organisms can reproduce sexually or asexually.

- **Sexual reproduction** involves <u>two</u> parents and produces genetically unique offspring. Each parent produces a sex cell (**gamete**), which fuse as part of sexual reproduction. This means that each parent contributes 50% of the genetic information to the offspring, and the offspring is *genetically unique*.
- **Asexual reproduction** involves only one parent and there is **no** fusion of gametes. As a result, there is **no** mixing of genetic information and the offspring are *genetically identical* to the parent (they are **clones** of their parent). No meiosis takes place (since there are no gametes); only mitosis is involved.

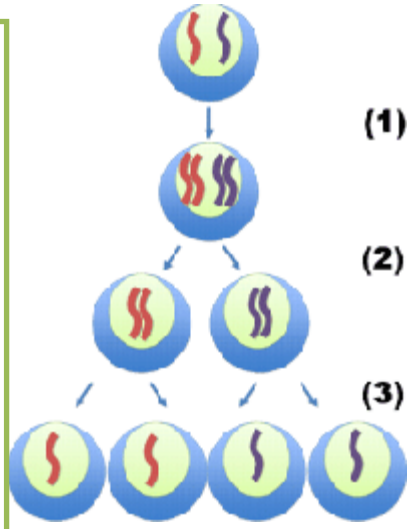
| Key Terms | Definitions |
|---|---|
| Reproduction | Making offspring. All organisms reproduce. |
| Offspring | Offspring is a generic term for children – it applies to any type of organism. |
| Gametes | Sex cells, such as pollen, egg cells, sperm cells. Gametes are produced by meiosis. |
| Meiosis | Type of cell division that produces gametes. Gametes are genetically unique (compare to *mitosis*, where genetically identical daughter cells are produced). |
| Fusion | The joining/fusing of sex cells in sexual reproduction. |
| Differentiation | The process of becoming a specialised cell. Specialised cells are the result of differentiation of **stem cells**. |

## Meiosis

You already know how mitosis is used to replace cells in the body. Meiosis is the other form of cell division, but quite different. Meiosis produces **<u>gametes</u>**, so it happens in **reproductive organs** (e.g. sperm cells are produced by meiosis in the testes; egg cells are produced by meiosis in the ovaries).

DNA in the nucleus of cells is arranged into structures called **chromosomes**. In all body cells, the chromosomes appear in pairs (in humans, there are 23 pairs, so 46 chromosomes altogether). However, in gametes, there are **half** the number of chromosomes of body cells, since they contain one from each chromosome pair (in humans, this means that gametes contain 23 chromosomes).

In meiosis, the DNA is replicated to start with (just like mitosis – step 1 in diagram). But then the cell divides **twice** – i.e. divides into *four* cells – so each cell ends up with **half** the genetic information: a <u>single set</u> of chromosomes. At stage 2 – the pairs are split up, then at stage 3 the copies of chromosomes are separated. The four cells produced are **gametes**, and all of them are **different** to each other – they are <u>genetically unique</u>.




## Fertilisation

Obviously, fertilisation only happens in sexual reproduction. The male and female gametes **fuse**. Their nuclei join together into one and the genetic information is combined. Consequently, you have 50% of your genetic information from your mother and 50% from your father. The cell that is produced has the full set of chromosomes (in pairs again) – the normal number is restored. Again, this is 46 chromosomes (23 pairs) in humans. The diagram shows this.

The new cell is ready to grow into an embryo. It does this through mitosis, increasing the number of cells. To be precise, each cell divides to make two cells. This means that a young embryo doubles the number of cells each 'round' of mitosis. After a ball of cells is produced, the cells start to **differentiate** – become specialised. So you are no longer just a blob.