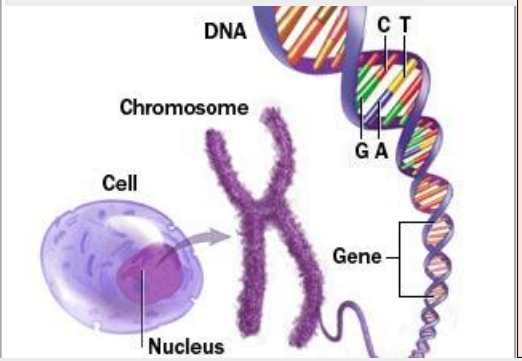
KS4 Knowledge organiser- Genetics

DNA is the **molecule** that carries our inheritance,. The **genome** of an organism is the entire genetic material of that organism. Our **DNA (deoxyribonucleic acid)** is a **polymer** of **nucleotides** that is made up of two double strands, forming a **double helix**. **DNA** is found within structures called **chromosomes.** A short section of **DNA** that controls a particular characteristic of an organism (i.e. eye colour or their blood group)is called a **gene**.

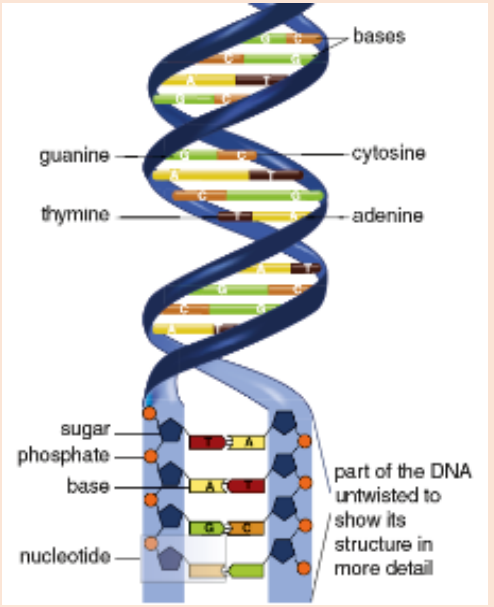


The **structure** of DNA was discovered in 1953, a molecule of DNA takes on the shape of a twisted ladder, called a **double helix**. The two "supports" of the ladder are made up of alternating **sugar molecules** and phosphate groups. Each "**rung**" of the **ladder** is made up of chemicals called **bases**, each base is attached to the sugar support structure of the DNA. The four bases are;

- **Adenine** (A)
- **Thymine** (T)
- **Cytosine** (C)
- **Guanine** (G)

The DNA molecule is made up of repeating units called **nucleotides.**

The **bases** of the DNA molecule make up the **genetic code.** They always pair up in the same way and are said to be **complementary**, A is always linked to T and C is always linked to G.

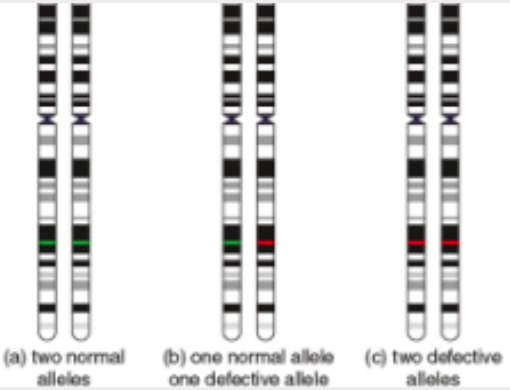


In 1990 a organisation called the **human genome project**, its aim was to map all of the genetic information found in the **chromosomes** of a human being. Their work was published in 2003. Their work has helped scientists understand more about diseases such as diabetes, cancer and heart disease. So far, around **4000 genes** have been found to be linked with human diseases. **Inherited diseases** such as cystic fibrosis are linked with just one gene. As a result of this information new forms of medicine such as **gene therapy** and **genome editing** are being developed to target faulty genes.

Within an individual's **genome** you will find the entire genetic makeup for that individual. Due to each cell having **pairs of chromosomes,** an individual can two different copies of a gene. These different copies are called **alleles.** What is expressed (physically seen, e.g. eye colour) is called the **phenotype.** The phenotype will be determined by what alleles are present. Alleles will either be always expressed **(dominant)** or sometimes expressed **(recessive).** Genetic diseases can be traced and predicted using an individual's genetic make-up. It is only with **two defective** alleles that a person will suffer from cystic fibrosis, if an individual has one normal and one defective they will not suffer with the disease but instead will be a **carrier**. This means that they could **pass** on the genes to any **offspring**. In order to represent inheritance of genes and diseases we can use genetic crosses. If we use cystic fibrosis as an example again, then a mother who suffers from cystic fibrosis must have a **double recessive allele** .We can represent this as **cc** (this is called a **genotype**). Someone who doesn't suffer from the disease can either be Cc (a carrier) or CC (no defective allele at all). To then work out the probability of the displaying this characteristic (this is called the **phenotype)** we use a **Punnett square.** Although the probability and ratios can be calculated, whether or not a particular phenotype will be expressed will be down to which sperm fertilises the egg.

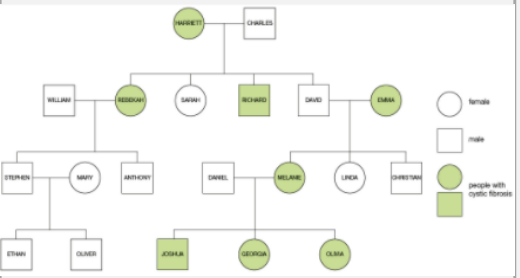
		Mother (cc) gametes	
		c	c
Father (Cc) gametes	C	Cc unaffected (but a carrier)	Cc unaffected (but a carrier)
	c	cc cystic fibrosis	cc cystic fibrosis

Cystic fibrosis is an inherited disorder. Those who suffer from the disease produce mucus that is thicker and stickier than normal, this can result in infections and difficulty breathing. If a sufferer has a lung transplant, this will not cure the disease as the body will continue to produce the mucus. Cystic fibrosis is caused by a defective **recessive allele.** It will only be caused if there is **no healthy allele present**.



Polydactyly is an inherited disorder resulting from a defective **dominant allele**. This means that you cannot be a carrier, if the defective allele is present, they will have the disorder. A sufferer will be born with extra fingers or toes, these will likely be removed at birth.

Most, but not all, **inherited diseases** are caused by **recessive alleles**. Some can be the result of a defective **dominant allele**, if this is the case then the presence of one allele will cause the **characteristic** to be seen in the **phenotype**. To see how the characteristic is inherited down a family we can use a family tree;



Mutations are changes to our DNA, they are happening **continuously** in our bodies. They can be a result of **radiation** or chemicals, or they can just happen **spontaneously**.

Although mutations can result in cancer and the death of an organism, they can also have very little effect. Because we have **two copies** of each gene, if one is faulty there is always a back up. This means that the protein produced by that gene can still be produced as normal. If the mutation occurs in one of the **bases** of a **DNA** molecule, then that gene may lead to another **amino acid** to be assembled into the protein, This could result in the **protein** being **shorter** or a **different shape** entirely, and if its an **enzyme** then it may no longer function.

Gregor Mendel established the basis of modern genetics. He investigated pea plants as there are a wide range of varieties available . For each characteristic he cross bred the plant with a plant of another breed. By doing this he proved the inheritance of characteristics as second and third generations displayed characteristics from the original plants.

Asexual reproduction is requires only one parent, unlike **meiosis.** The offspring will be a clone, with new cells being produced by **mitosis. Flowering plants** are able to reproduce both sexually and asexually. Despite sexual reproduction offering genetic variation and better chances of survival through natural selection, asexual reproduction still has benefits

- **There is no need to find a mate as it requires less energy.**
- **This organism can produce a large quantity of offspring when the conditions are favourable.**

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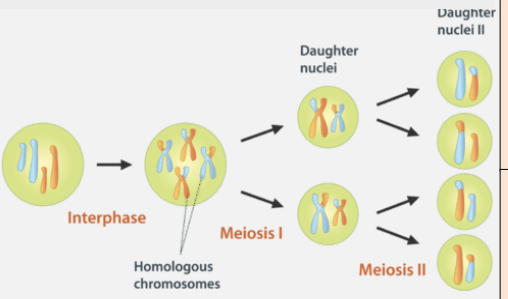
Meiosis is a type of cell division that occurs during the formation of sex cells. During meiosis;

- Four **gametes** are produced from one parent cell.
- Each **gamete** has **half** the number of **chromosomes** than a regular cell (23 instead of 46).

The DNA of each chromosome is copied, just like **Mitosis,** but it is then divided twice, so the number is halved.



When the **gametes** fuse during fertilisation, the normal number is restored. One of each **chromosome pair** will be from the mother and one from the father, but it is completely random which goes into the egg or sperm. Unlike mitosis, during meiosis, there is an **exchange** of genetic material, ensuring **genetic variation**. Each of the gametes will also carry one of two sex chromosomes, which will determine the sex of any children.

Father (XY) gametes	Mother (XX) gametes: X	Mother (XX) gametes: X
X	XX female	XX female
Y	XY male	XY male

When a couple use in-vitro fertilisation, several embryos are usually produced. A few cells are tested for defective alleles, this is called **embryo screening.** In families where **genetic disorders** are common, once screened, it is only then that the decision is made to implant it into the mother. This raises several **ethical**, **social** and **economic** issues. This is because embryos that aren't used will be frozen, but eventually they could be **destroyed**. Another worry that is embryo screening will lead to designing babies for social reasons, such as choosing gender or skin colour etc.

Genes code for the assembly of **proteins**, these are **essential** for the daily tasks that every cell are required to carry out. Every protein is made up of different combinations of only **20 amino acids**. The four-letter base code (**A,C,T,G**) works in threes, with each 3 letter code (or **triplet**) coding for a specific **amino acid**. Proteins are assembled on a **ribosome** within a cell, with the sequence of bases in a gene acting as a messenger molecule.