Chromosomes, genes, and proteins

Chromosome structure

During the phase of cell cycle known as interphase, chromosomes are in the form of long, very thin threads, which cannot be seen with a simple microscope. As the nucleus prepares to divide, these threads undergo repeated coiling and become much shorter and thicker. When stained they are clearly visible even at low microscope magnification.

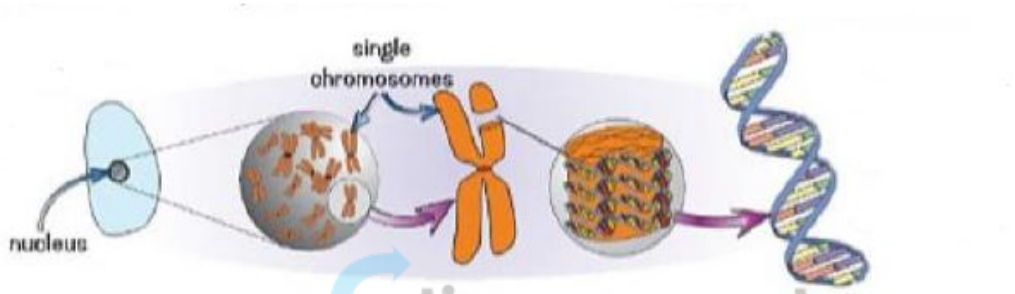
Eukaryotic cells contain chromosomes surrounded by a nuclear envelope. The chromosomes of eukaryotic cells are associated with proteins. Each chromosome contains a single molecule of DNA along with the associated proteins. Some of these proteins are structural and others regulate the activities of the DNA.

DNA

Chromosomes are really long molecules of DNA

- DNA stands for deoxyribonucleic acid, It’s the chemical that all of the genetic material in a cell is made up from.
- It contains coded information – basically all the instructions to put an organism together and make it work.

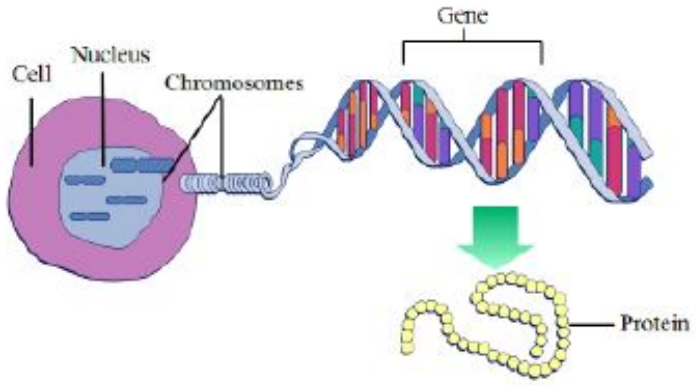
- So it’s what’s in your DNA that determines what inherited characteristics you have.
- DNA is found in the nucleus of animal and plant cells, in really long structures called chromosomes.
- Chromosomes normally come in pairs.
- DNA is a polymer. It’s made up of two strands coiled together in the shape of a double helix.



A DNA molecule with a double helix structure (a double-stranded spiral)

A gene codes for a specific protein

- A gene is a small section of DNA found on the chromosome.
- Each gene codes for a particular sequence of amino acids which are put together to make a specific protein.
- Only 20 amino acids are used, but they make up thousands of different proteins.
- Genes simply tell cells in what order to put the amino acids together.
- DNA also determines what proteins the cell produces e.g. haemoglobin, keratin.
- That in turn determine what type of cell it is e.g. red blood cell, skin cell.



A part of a DNA molecule coding for one protein is called a gene

Key definitions

Chromosome: A thread of DNA, made of genes.
Gene: A section of DNA, which codes for the formation of a protein controlling a specific characteristic of the organism.

Every organisms has a genome

1. Genome is just the fancy term for the entire set of genetic material in an organism.
2. Scientists have worked out the complete human genome.
3. Understanding the human genome is a really important tool for science and medicine for many reasons.
    - It allows scientists to identify genes in the genome that are linked to different types of disease.
    - Knowing which genes are linked to inherited diseases could help us

to understand them better and could help us to develop effective treatments for them.

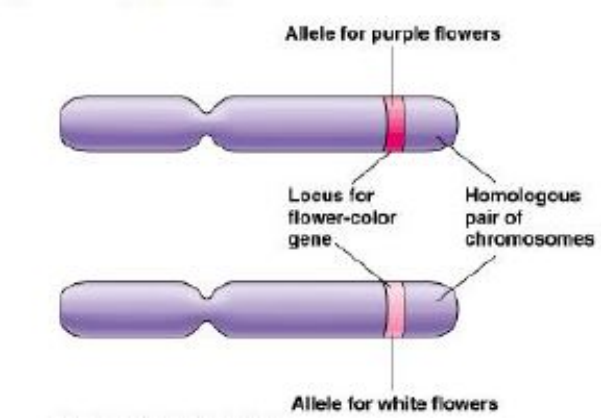
- Scientists can look at genomes to trace the migration of certain populations of people around the world. All modern humans are descended from a common ancestor who lived in Africa, but humans can now be found all over the planet. The human genome is mostly identical in all individuals, but as different populations of people migrated away from Africa, they gradually developed tiny differences in their genomes. By investigating these differences, scientists can work out when new populations split off in a different direction and what route they took.

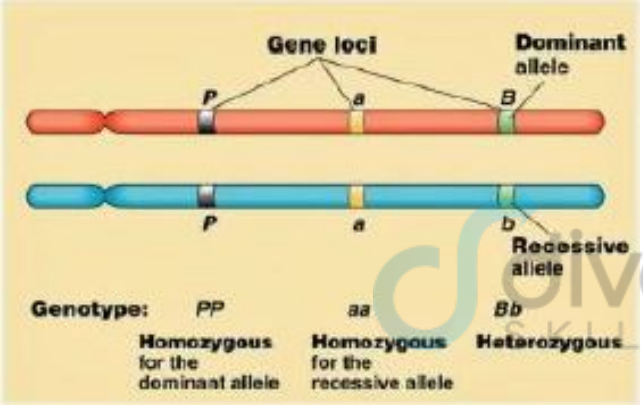
Alleles

When the chromosomes are in pairs, there may be a different form (allele) of the gene on each chromosome.

Alleles for the same gene can be;

- Dominant – always affect the phenotype
- Recessive – never affect the phenotype in the presence of a dominant allele
- Co-dominant – affect the phenotype equally in the presence of another co-dominant allele





Key definitions

Phenotype: The characteristics visible in an organism, controlled by the genotype, e.g. a tall plant or a dwarf plant.

Genotype: The genetic make-up of an organism, e.g. Tt, where T and t are alleles of a gene.

Homozygous: Having a pair of identical alleles controlling the same characteristics, e.g. TT, where T=tall. The organism will be pure-breeding for that characteristics.
Heterozygous: Having a pair of dissimilar alleles for a characteristic, e.g. Tt.
Dominant: A gene, e.g. T, that always shows in the phenotype of an organism whether the organism is heterozygous (Tt) or homozygous (TT).
Recessive: A gene, e.g. t, that only has an effect on the phenotype when the organism is homozygous (tt).

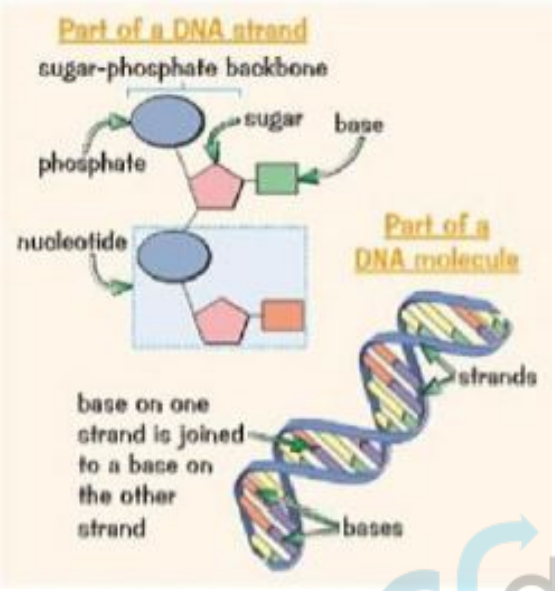
The structure of DNA and protein synthesis

DNA molecules contain a genetic code that determines which proteins are built.

DNA is made up of nucleotides

- DNA strands are polymers made up of lots of repeating units called nucleotides.
- Each nucleotide consists of one sugar molecule, one phosphate molecule and one 'base'.
- The sugar and phosphate molecules in the nucleotides form a 'backbone' to the DNA strands. The sugar and phosphate molecules alternate. One of the four different bases – A, T, C or G – joins to each sugar.
- Each base links to a base on the opposite strand in the helix.
- A always pairs up with T, and C always pairs up with G. This is called complimentary base pairing.

- It’s the order of bases in a gene that decides the order of amino acids in a protein.



- Each amino acid is coded for by a sequence of three bases in the gene.
- The amino acids are joined together tho make various proteins, depending on the order of the gene’s bases.
- There are parts of DNA that don’t code for proteins. Some of these non-coding parts switch genes on and off, so they control whether or not a gene is expressed (used to make a protein).

mRNA carries the code to the ribosomes

- Proteins are made in the cell cytoplasm on tiny structures called ribosomes.
- To make proteins, ribosomes use the code in the DNA. DNA is found in the

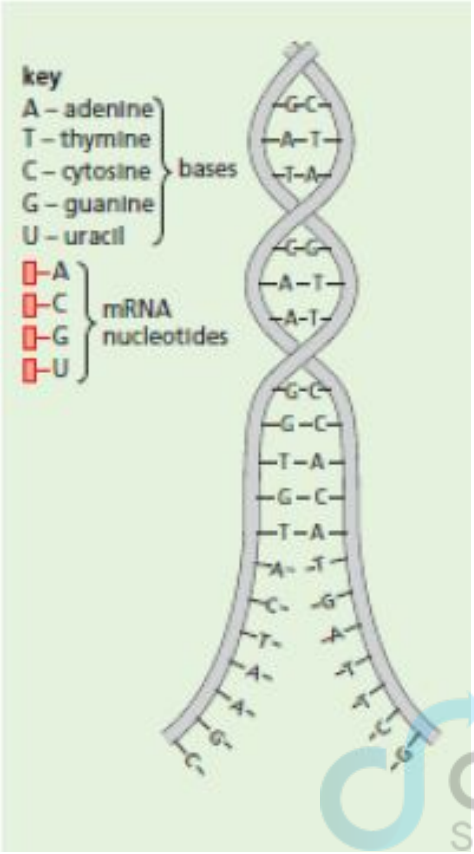
cell nucleus and can't move out of it because it's really big. So the cell needs to get the code from the DNA to the ribosome.

- This is done using a molecule called mRNA – which is made by copying the code from DNA. The mRNA acts as a messenger between the DNA and the ribosome – it carries the code between the two.
- The correct amino acids are brought to the ribosomes in the correct order by carrier molecules.

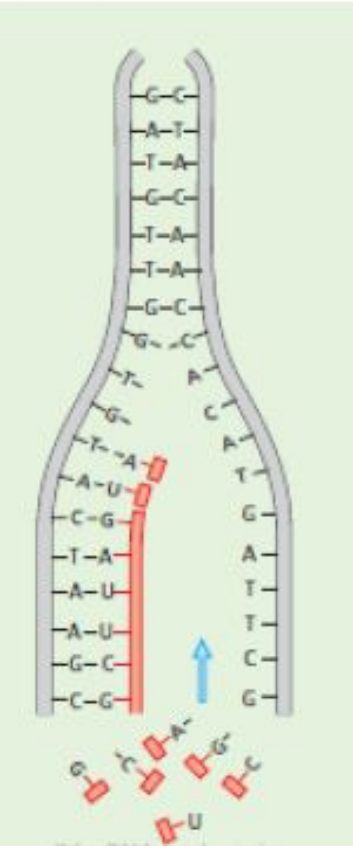
The manufacture of proteins in cells

DNA molecules remain in the nucleus, but the proteins they carry the codes for are needed elsewhere in the cell. A molecule called messenger RNA (mRNA) is used to transfer the information from the nucleus. It is much smaller than a DNA molecule and is made up of only one strand. Another difference is that mRNA molecules contain slightly different bases (A, C, G and U). Base U is uracil. It attaches to the DNA base A.

To pass on the protein code, the double helix of DNA unwinds to expose the chain of bases. One strand acts as template. A messenger RNA molecules is formed along part of this strand, made up of a chain of nucleotides with complementary bases to a section of the DNA strand see Figure below.



(a) The DNA helix unwinds: the strands separate, exposing the bases



(b) mRNA nucleotides to the exposed DNA bases to form a mRNA molecule

Formation of messenger RNA

The mRNA molecule carrying the protein code then passes out of the nucleus, through a nuclear pore in the membrane. Once in the cytoplasm it attaches itself to a ribosome. Ribosomes make proteins. The mRNA molecule instructs the ribosome to put together a chain of amino acids in a specific sequence, thus making a protein. Other mRNA molecules will carry codes for different proteins.

Some proteins are made up of relatively small number of amino acids. As stated, insulin is a chain of 51 amino acids. On the mRNA molecule each amino acid is coded by a sequence of three bases (a triplet), so the mRNA molecule coding for

insulin will contain 153 bases. Other protein molecules are much bigger: haemoglobin in red blood cells is made of 574 amino acids.

Proteins have many different functions

When a chain of amino acids has been assembled, it folds into a unique shape which allows the protein to perform the task it's meant to do. Here are a few examples of types of protein:

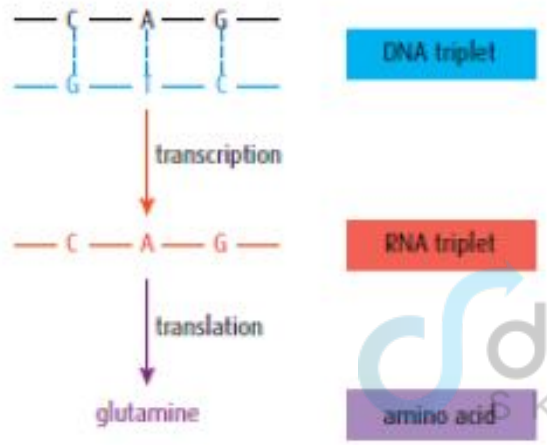
- **Enzymes** – act as biological catalysts to speed up chemical reactions in the body.
- **Hormones** – used to carry messages around the body. E.g. insulin is a hormone released into the blood by the pancreas to regulate the blood sugar level.
- **Structural proteins** – are physically strong. E.g. collagen is a structural protein that strengthens connective tissues (like ligaments and cartilage).

Chromosomes, genes and mutations

A DNA molecule comprises a pair of strands, each strand consisting of a linear sequence of nucleotides, held together by weak bonds between the bases. This linear sequence of bases contains the genetic code in the form of triplets of bases. A gene is a particular section of DNA strand that, when transcribed and translated, forms a specific polypeptide.

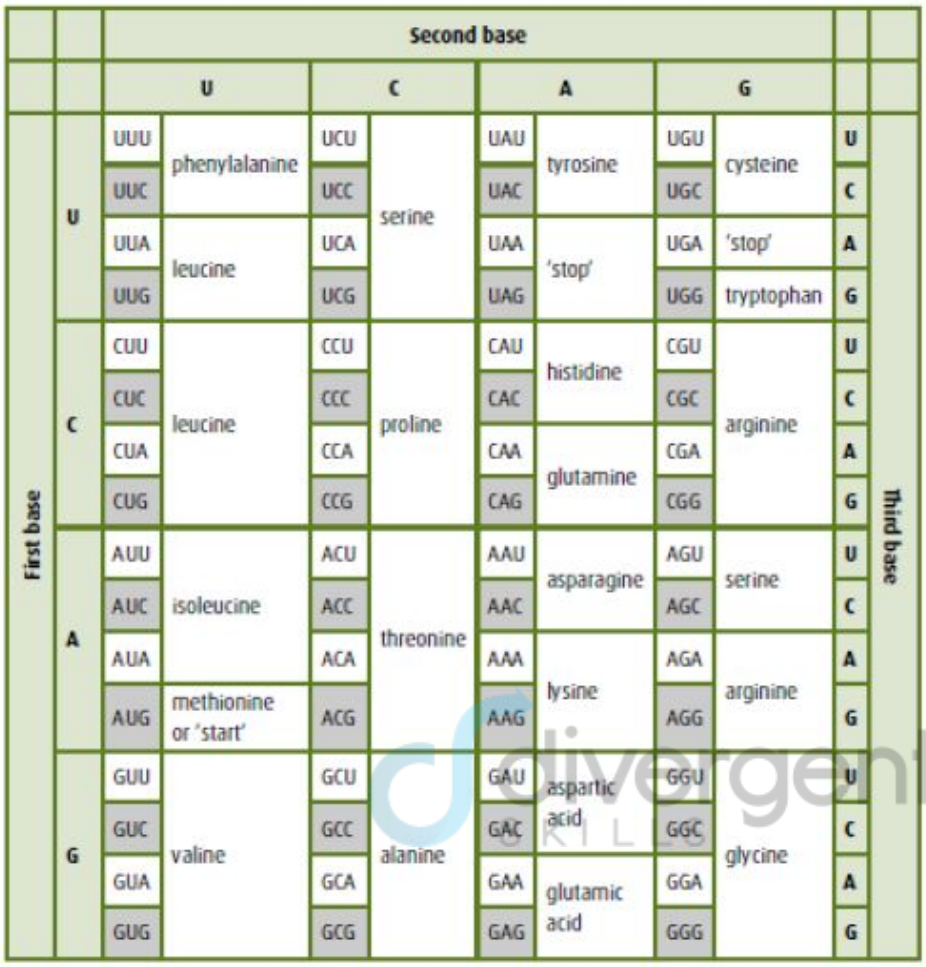
A triplet of bases in the DNA molecule is transcribed into a triplet of bases in the mRNA molecule, which is then translated into a specific amino acid as shown in Figure.

The process of DNA replication is complex and mistakes sometimes occur – a nucleotide may be left out, an extra one may be added, or the wrong one inserted. These mistakes are known as gene mutations. The insertion of an incorrect nucleotide is called a base substitution mutation. When the DNA containing an incorrect nucleotide is transcribed and translated, errors may occur in the polypeptide produced.



The base sequence in DNA is decoded via transcription and translation

Table below shows the amino acids that are specified by different mRNA codons. Most amino acids are coded for by more than one codon and so many substitution mutations have no effect on the final polypeptide that is produced. For example, a mutation in the DNA triplet CCA into CCG would change the codon in the mRNA from GGU to GGC but it would still result in the amino acid glycine being placed in a polypeptide. Some substitution mutations, however, do have serious effects and an important human condition that results from a single base substitution is sickle-cell anemia.



First base		Second base U		Second base C		Second base A		Second base G		Third base
U		UUU	phenylalanine	UCU	serine	UAU	tyrosine	UGU	cysteine	U
		UUC		UCC		UAC		UGC		C
		UUA	leucine	UCA		UAA	'stop'	UGA	'stop'	A
		UUG		UCG		UAG		UGG	tryptophan	G
C		CUU	leucine	CCU	proline	CAU	histidine	CGU	arginine	U
		CUC		CCC		CAC		CGC		C
		CUA		CCA		CAA	glutamine	CGA		A
		CUG		CCG		CAG		CGG		G
A		AUU	isoleucine	ACU	threonine	AAU	asparagine	AGU	serine	U
		AUC		ACC		AAC		AGC		C
		AUA		ACA		AAA	lysine	AGA	arginine	A
		AUG	methionine or 'start'	ACG		AAG		AGG		G
G		GUU	valine	GCU	alanine	GAU	aspartic acid	GGU	glycine	U
		GUC		GCC		GAC		GGC		C
		GUA		GCA		GAA	glutamic acid	GGA		A
		GUG		GCG		GAG		GGG		G

To show the amino acids and their associated mRNA codons

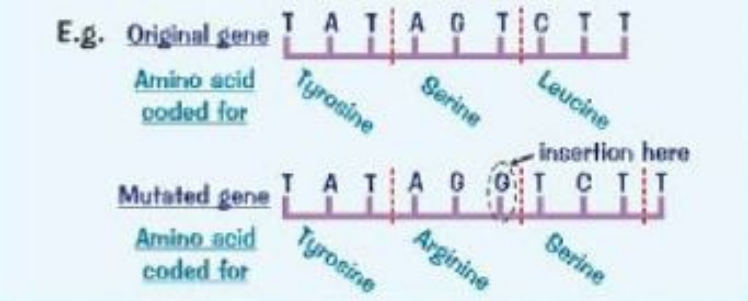
There are different type of Mutation, for example…..

Insertions

- Insertions are were a new base is inserted into the DNA base sequence where it shouldn't be.
- You should remember that every three bases in a DNA base sequence

codes for a particular amino acid.

- An insertion changes the way the groups of three bases are ‘read’, which can change the amino acids that they code for.
- Insertions can change more than one amino acid as they have knock-on effect on the bases further on in the sequence.

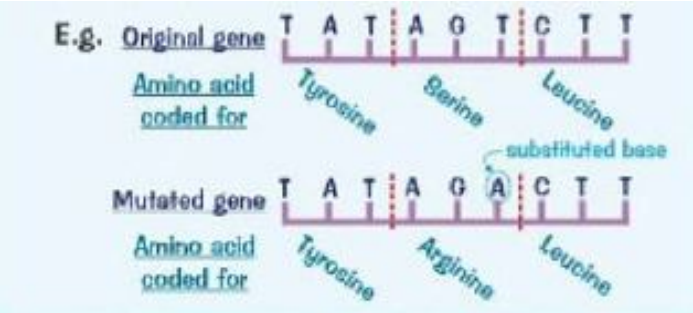


Deletions

- Deletions are when a random base is deleted from the DNA base sequence.
- Like insertions, they change the way that the base sequence is ‘read’ and have knock-on effects further down the sequence.

Substitutions

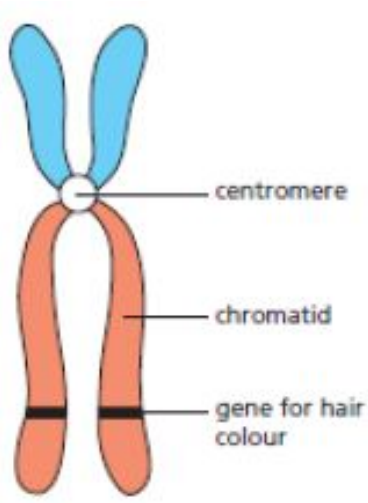
Substitution mutations are when a random base in the DNA base sequence is changed to a different base.



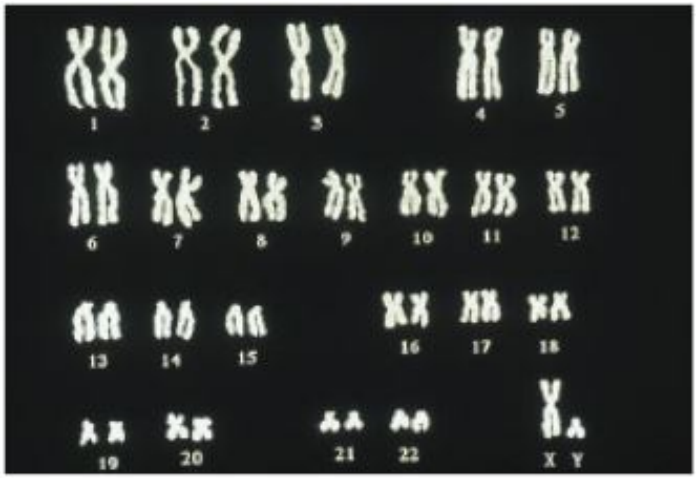
Number of chromosome

Each chromosome has certain characteristics when ready to divide: there are two chromatids, joined at one point called a centromere see Figure below. Each chromatid is a string of genes, coding for the person's characteristics. The other chromatid carries the same genes in the same order.

A human body (somatic) cell nucleus contains 46 chromosomes. These are difficult to distinguish when packed inside the nucleus, so scientists separate them and arrange them according to size and appearance. The outcome is called a karyotype see Figure below. There are pairs of chromosomes (they come from a diploid cell). Because the chromosomes are in pairs, the diploid number is always an even number. The karyotype of a sperm cell would show 23 single chromosomes (they come from a haploid cell). The only pair that do not necessarily match is chromosome pair 23: the 'sex chromosomes'. The Y chromosome is much smaller than the X chromosome.

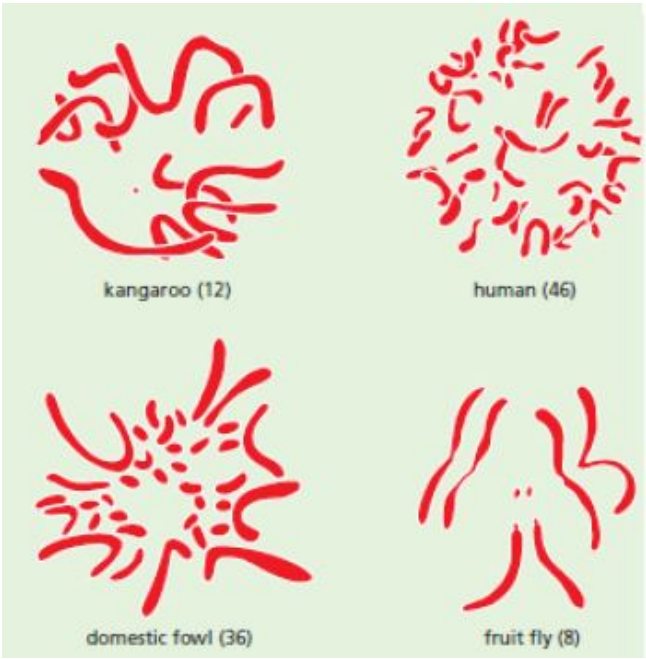


Structure of a chromosome



Human karyotype

The chromosomes have different shapes and sizes and can be recognised by a trained observer. There is a fixed number of chromosome in each species. Human body cells each contain 46 chromosomes, mouse cells contain 40 and garden pea cell 14.



Chromosomes of different species.

The number of chromosomes in a species is the same in all of its body cells. There are 46 chromosomes in each of your liver cells, in every nerve cell, skin cell and so on.

The chromosomes are always in pairs, e.g. two long ones, two short ones, two medium ones. This is because when the zygote is formed, one of each pair comes from the male gamete and one from the female gamete. Your 46 chromosomes consist of 23 from your mother and 23 from your father. The chromosomes of each pair are called homologous chromosomes.

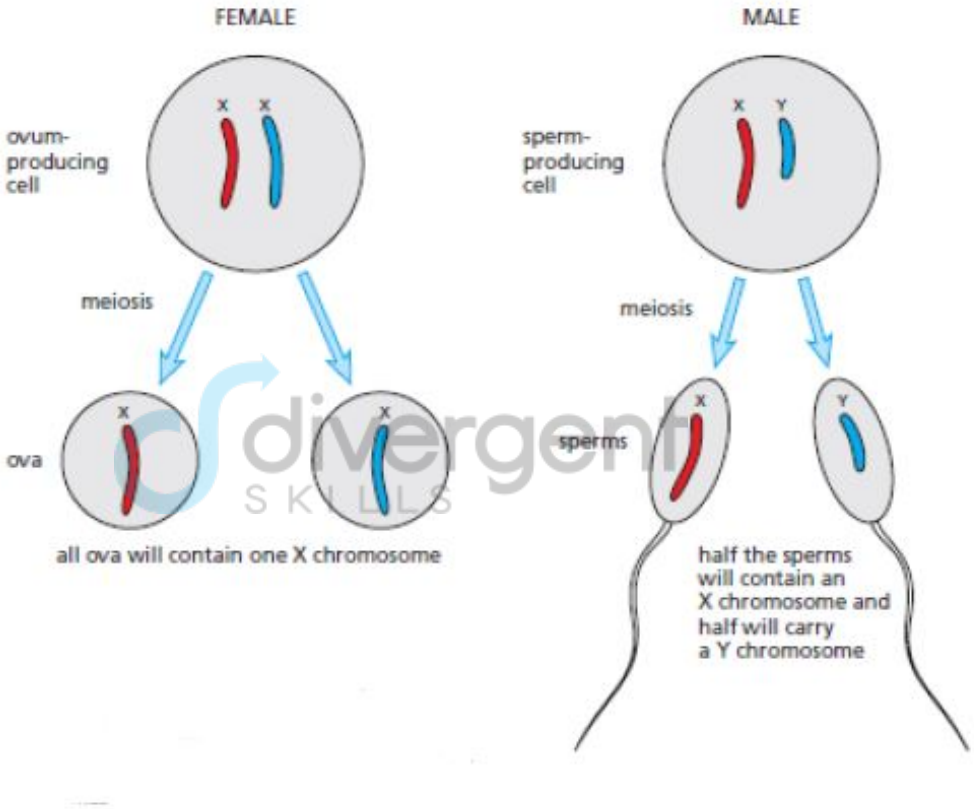
Key definition

Haploid nucleus: A nucleus containing a single set of unpaired chromosomes, e.g. in sperm and ova (eggs). In humans, the haploid number is 23.
Diploid nucleus: A nucleus containing pairs of chromosomes, e.g. in somatic (body) cells. In humans the diploid number is 46.

The inheritance of sex

Whether you are male or female depends on the pair of chromosomes called the 'sex chromosomes'. In females, the two sex chromosomes, called the X chromosomes, are the same size as each other. In males, the two sex chromosomes are of different sizes. One corresponds to the female sex X chromosome and is called the X chromosome. The other is smaller and is called the Y chromosome. So the female cells contain XX and male contains XY.

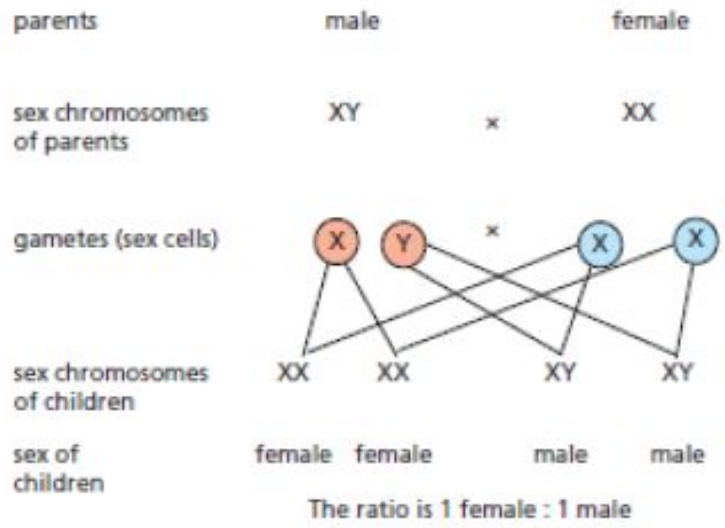
A process called meiosis takes place in the female's ovary. It makes gametes: sex cells, which have half the normal number of chromosomes. During the process, each ovum receives one of the X chromosomes, so all the ova are the same for this. Meiosis in the male's testes results in 50% of the sperms getting an X chromosome and 50% getting a Y chromosome see Figure below.



Determination of sex. Note that: (i) only the X and Y chromosomes are shown (ii) details of meiosis have been omitted (iii) in fact, four gametes are produced in each case, but two are sufficient to show the distribution of X and Y chromosomes

If an X sperm fertilises the ovum, the zygote will be XX and will grow into a girl. If a Y sperm fertilises the ovum, the zygote will be XY and will develop into a boy. There is an equal chance of an X or Y chromosome fertilising an ovum, so the numbers of girl and boy babies are more or less the same.

Figure below shows how sex is inherited.



Determination of sex