Topic 3: Voice of the Genome

Sexual Reproduction and Meiosis

Gametes

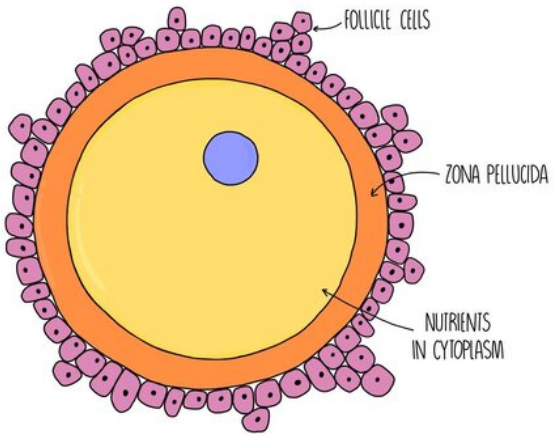
Gametes are **sex cells** (the sperm and egg in humans). Gametes are **haploid** which means they contain half the number of chromosomes as the rest of the cells which make up our body. This means that when two gametes fuse during sexual reproduction, the fertilised egg (called a **zygote**) contains the full number of chromosomes i.e. it is **diploid**. In humans, the diploid number of chromosomes is 46 (23 pairs), which means that gametes contain just 23 chromosomes.

During sexual reproduction, the nucleus of the sperm cell fuses with the nucleus of the egg cell - this fusion of nuclei is called **fertilisation**.

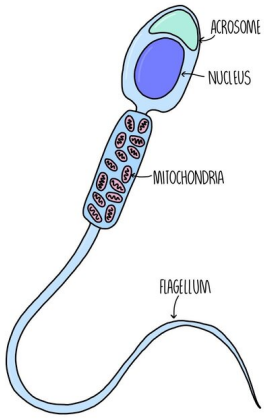
Adaptations of gametes

Egg cells are specialised for fertilisation in the following ways:

- Cytoplasm contains **nutrients** for growth of the developing embryo.
- There is an outer layer called the **zona pellucida** - this **changes** after fertilisation so that **no further sperm can penetrate**.
- **Follicle cells** outside the zona pellucida form a **protective coating** around the egg cell.



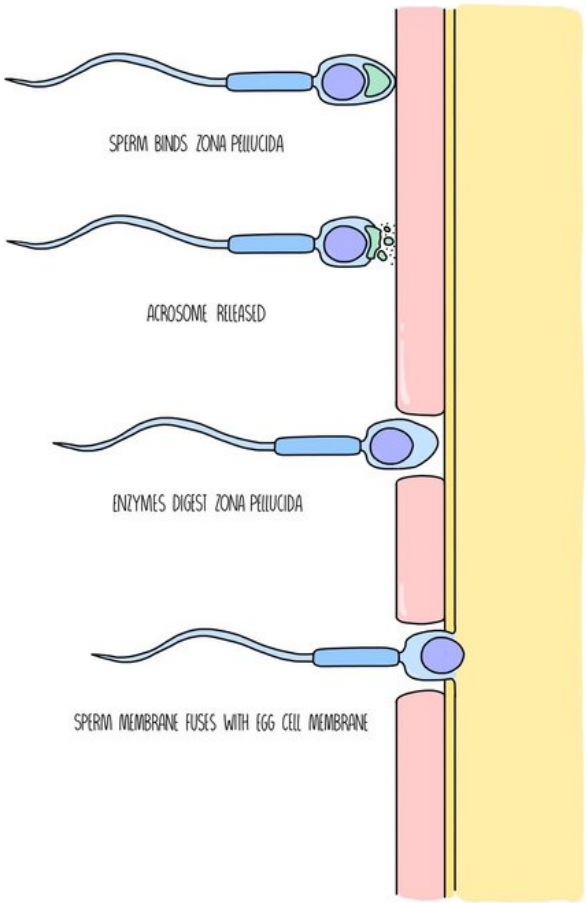
Sperm cells are also specialised to carry out fertilisation:



- Overall **streamlined shape** for faster swimming.
- Tail (**flagellum**) enables the sperm to swim.
- Contain lots of **mitochondria** to provide ATP for movement.
- The head of the sperm contains the **acrosome** - this is filled with digestive enzymes which break down the egg's zona pellucida and allow the sperm to reach the egg cell membrane.

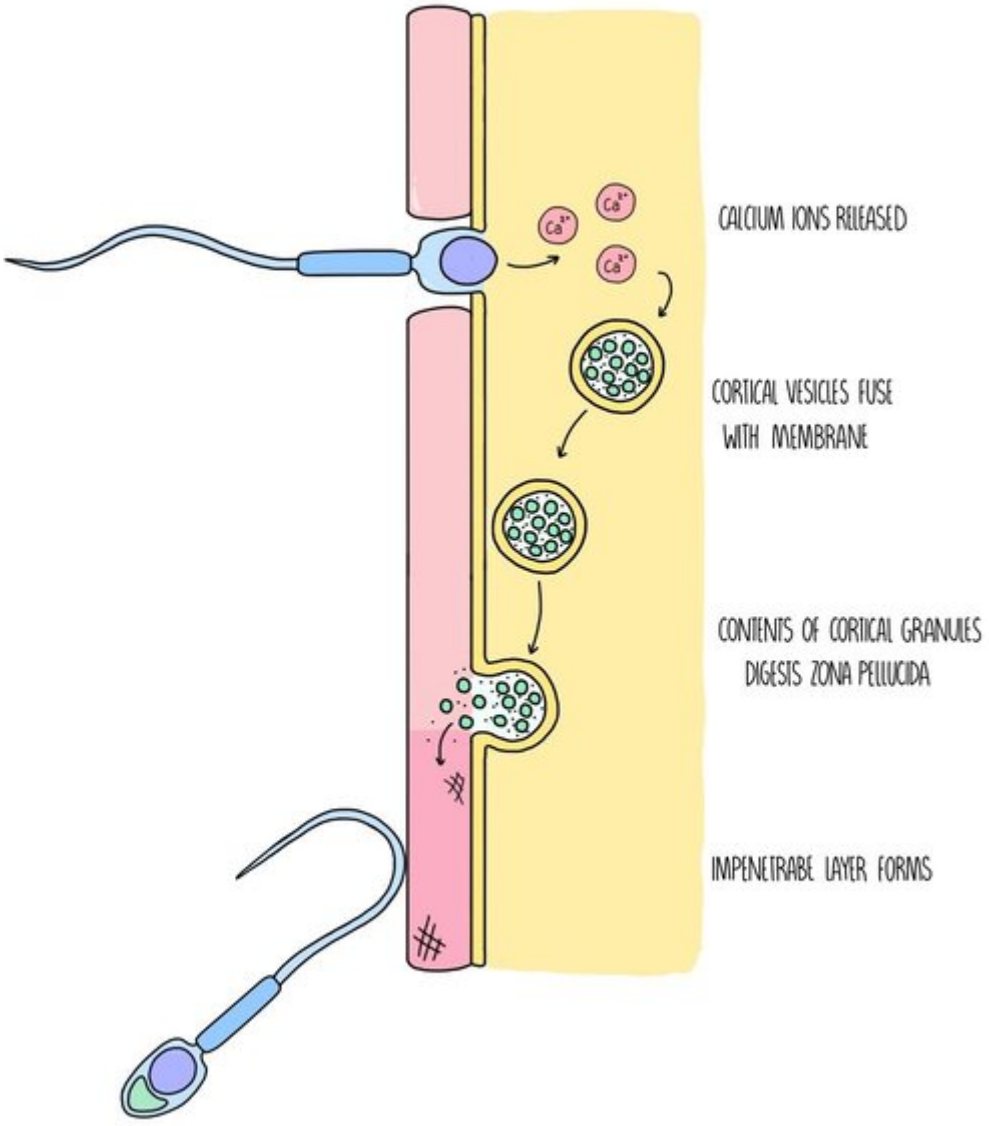
The Acrosome Reaction

When a sperm reaches the egg cell, it binds to its outer layer (the **zona pellucida**) through attachment to sperm-binding proteins. Once bound, it releases the **digestive enzymes** that are contained in the **acrosome**. These enzymes digest a tunnel through the zona pellucida so that the sperm can reach the plasma membrane of the egg cell. The **plasma membranes** of the sperm cell and the egg cell **fuse** and the sperm releases its nucleus into the egg cell cytoplasm. The nuclei of the sperm and egg fuse - **fertilisation** has taken place and a **zygote** is formed.



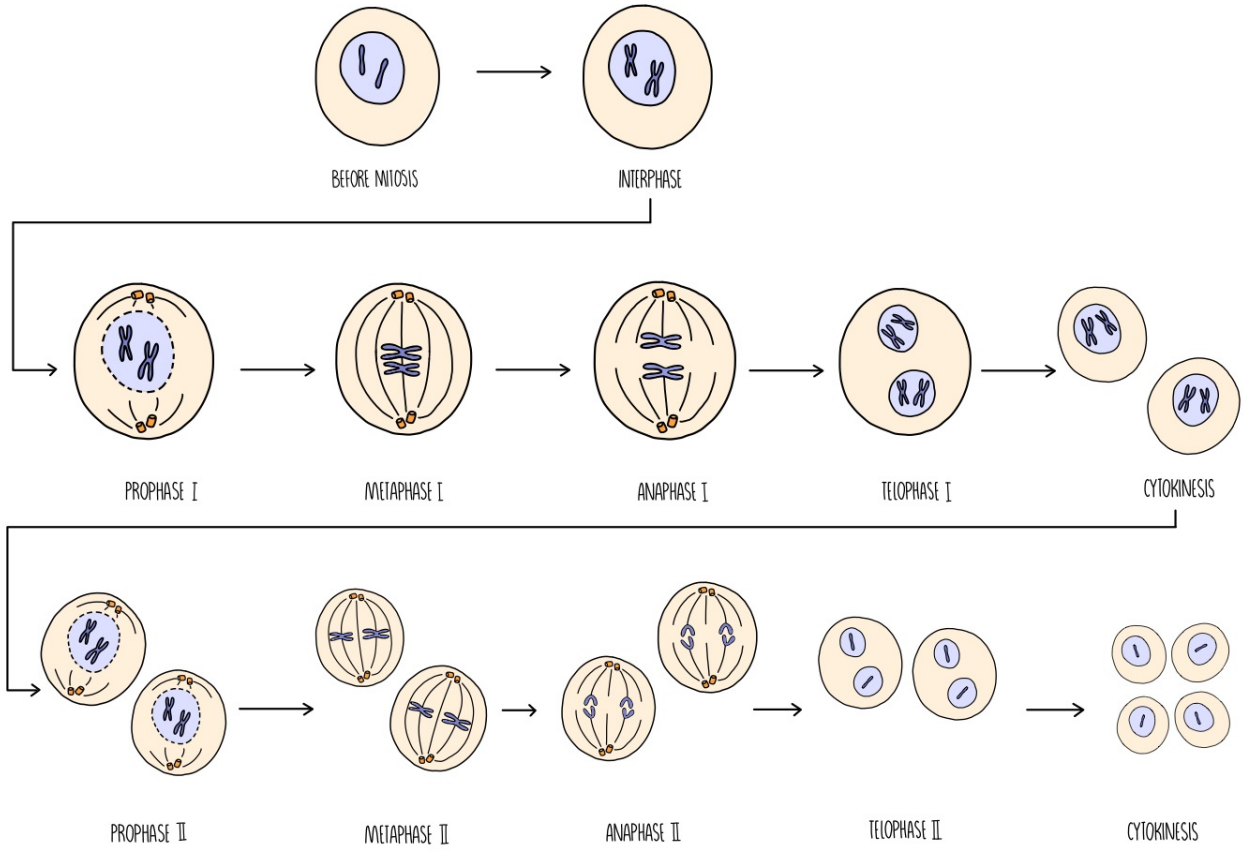
The Cortical Reaction

The fusion of the sperm cell and egg cell membranes triggers the release of **calcium ions** which stimulates **vesicles** containing **cortical granules** to move and fuse with the egg cell membrane. The cortical granules are released into the **zona pellucida**, which continue breaking down the zona pellucida, removing the remaining sperm-binding proteins so that **no further sperm** can bind. Other substances within the cortical granules produce a new outer layer which is **thick and impenetrable** to sperm cells. This process prevents multiple sperm cells from fertilising the egg, which would result in the zygote having an abnormal number of chromosomes.



Meiosis

Meiosis is the type of cell division which produces **gametes** for **sexual reproduction**. Unlike mitosis, the daughter cells are **genetically different** from the parent cell and contain just **half** the number of chromosomes (i.e. they are **haploid**). When two haploid gametes join during fertilisation, a **diploid** cell called a **zygote** is formed. Meiosis involves **two rounds** of cell division which are referred to as **meiosis I** and **meiosis II**.



It takes place in the following stages:

Meiosis I

- **Interphase**: the **DNA replicates** so there are now **two identical copies** of each chromosome (referred to as **chromatids**).
- **Prophase I**: chromatids **condense** and arrange themselves into **homologous pairs** (called bivalents). **Crossing over** occurs (see below). The **nuclear envelope disintegrates** and **spindle fibres form**.
- **Metaphase I**: homologous chromosomes line up along the **equator** and **attach to the spindle fibre** by their centromeres.

- **Anaphase I**: homologous chromosomes are **separated**
- **Telophase I**: chromosomes reach **opposite poles** of the cell. **Nuclear envelope reforms** around the chromosomes. **Cytokinesis** results in the formation of two daughter cells.

Meiosis II

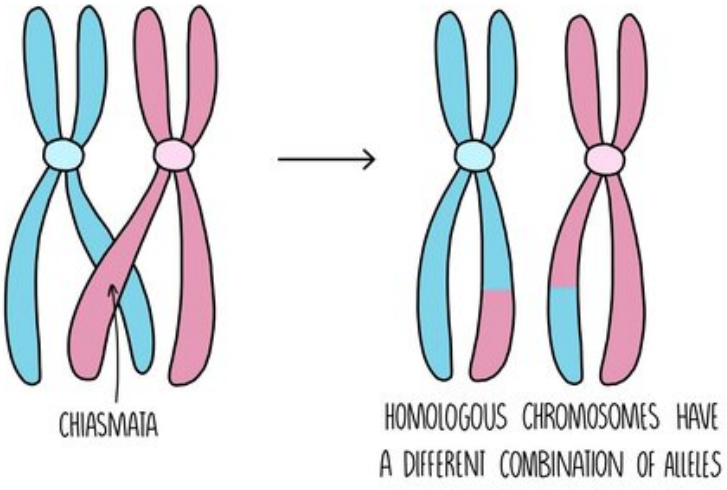
- **Prophase II**: chromosomes **condense**, **nuclear envelope disintegrates** and **spindle fibres form**.
- **Metaphase II**: chromosomes **attach to the spindle fibre** by their centromeres.
- **Anaphase II**: sister chromatids are **separated**.
- **Telophase II**: chromatids reach **opposite poles** of the cell. **Nuclear envelope reforms** and **cytokinesis** takes places. **Four genetically unique daughter cells** are produced.

Meiosis increases genetic variation

From an evolutionary point of view, it is important that organisms produce offspring that show as much **genetic variation** as possible. Imagine if a mother duck gave birth to a group of ducklings that were all had very similar genes - these ducklings will all be **equally vulnerable** to the same diseases and other threats to their survival. Meiosis increases genetic variation in two ways - **crossing over** and **independent assortment**.

Crossing Over

During prophase I of meiosis, a process called crossing over occurs. This is when the **homologous chromosomes** move towards each other and **exchange genetic material**. A chromatid from the maternal chromosome becomes twisted around the paternal chromosome and they connect through a structure called the **chiasmata**. Pieces of chromosomes are **exchanged** and the chromatids separate, forming chromosomes with **different combinations of alleles**.



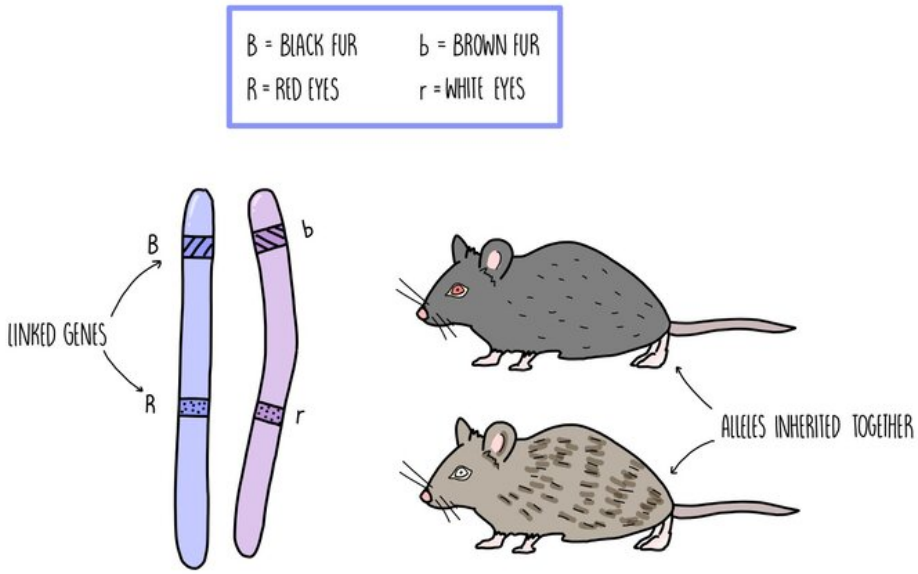
Independent assortment

Depending on the **order** in which chromosomes line up along the **equator** of the cell during **metaphase**, **different combinations** of chromosomes will end up in each gamete. The way in which the chromosomes align themselves on the spindle fibre is completely **random**, resulting in a huge number of possibilities of chromosomal combinations in the gametes.

Linked genes

The position of a gene on a chromosome is called its **locus**. If the loci of two different genes are on the **same chromosome**, they are likely to be **inherited together** and are said to be **linked**. The only way that the genes will not be inherited together is if crossing over separates them during **meiosis** (the **chiasmata** would have to form between the two genes). The **closer the loci** of the two genes, the **less likely** this is to happen and the higher the probability that the genes will be **inherited together**. This means that any offspring will probably **express both phenotypes** together than either phenotype separately.

The image below shows an example of gene linkage in rats. If the genes for coat colour and eye colour are located on the **same chromosome,** they will be **inherited together** and the offspring will show **both phenotypes together**. For example, there will be rats with both black fur and red eyes, and with both brown fur and white eyes but not many with black fur + white eyes (or likewise, brown fur + red eyes). To get these combinations of phenotypes, **crossing over** must have occurred between the homologous chromosomes to cause the alleles to end up on the same chromosome and be inherited together.



Sex Linkage

Genes which are **located on one of the sex chromosomes** (X or Y) are said to be **sex-linked** and their expression will depend on whether the offspring is male (**XY**) or female (**XX**). The Y chromosome is much **smaller** than the X chromosome, so most alleles are carried on the X chromosome (they are **X-linked**). Men only have **one X chromosome** which means that they will **only inherit one allele** for these genes, compared to **women** who will inherit **two**. This means that if men inherit a **recessive allele** (which causes disease) for a gene found on the X-chromosome, they will have the disease. Women who inherit the recessive allele will just be a **carrier**, since they have another X chromosome with the dominant, functioning allele. For women to have X-linked diseases, they must **inherit two disease alleles** (they will have a homozygous recessive phenotype). Examples of sex linked disorders include **haemophilia** and **red-green colour blindness**.