

Edexcel (A) Biology A-level

Topic 3: Voice of the Genome

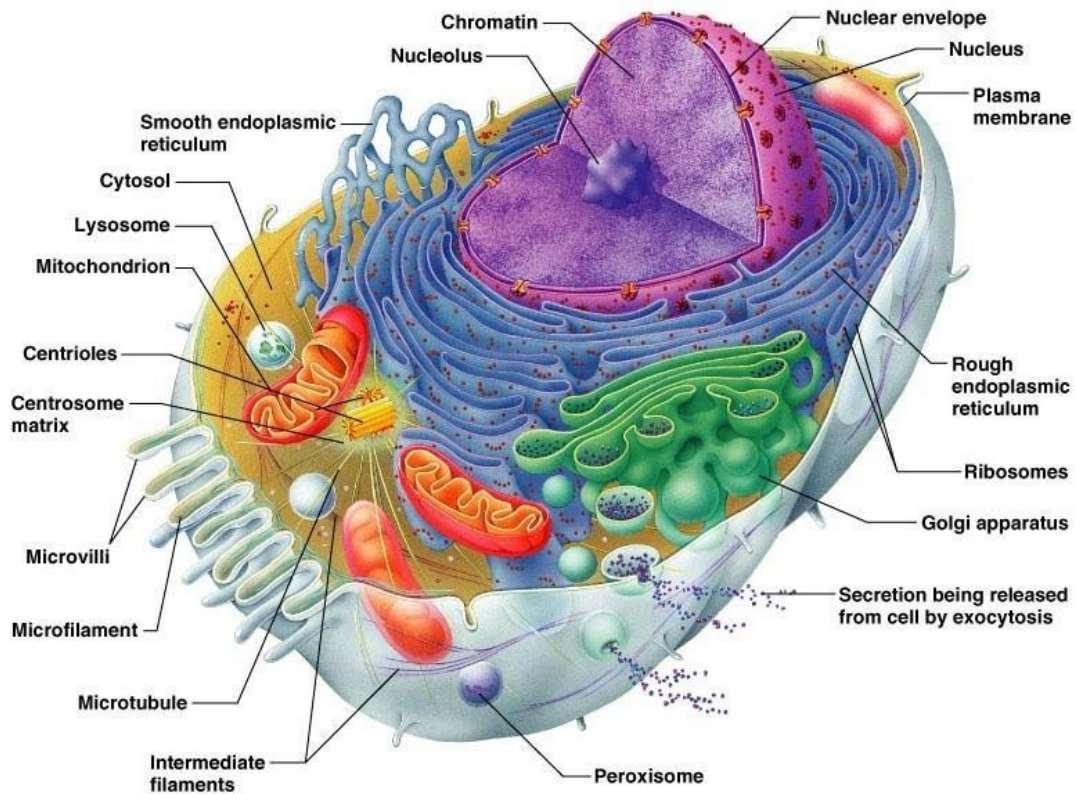
Notes



Cell Structure

All living organisms are made of cells and share some common features. In multicellular organisms, cells are organised into **tissues**, tissues into **organs** and organs into **organ systems**. There are several different types of cells; some of them share common features. Humans are made up of **eukaryotic cells**, which contain a **nucleus** and **membrane-bound organelles**. A more detailed structure of a cell, called the **ultrastructure**, can be observed using a microscope.

Ultrastructure of eukaryotic cells:



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- Nucleus – surrounded by a **double membrane** called the envelope, which contains pores enabling molecules to enter and leave the nucleus. The nucleus also contains **chromatin**, and a **nucleolus**, which is the site of **ribosome production**.
- Rough Endoplasmic Reticulum – a series of **flattened sacs enclosed by a membrane** with ribosomes on the surface. The RER **folds and processes proteins** made on the ribosomes.
- Smooth Endoplasmic Reticulum – a system of **membrane-bound sacs**. The SER **produces and processes lipids**.

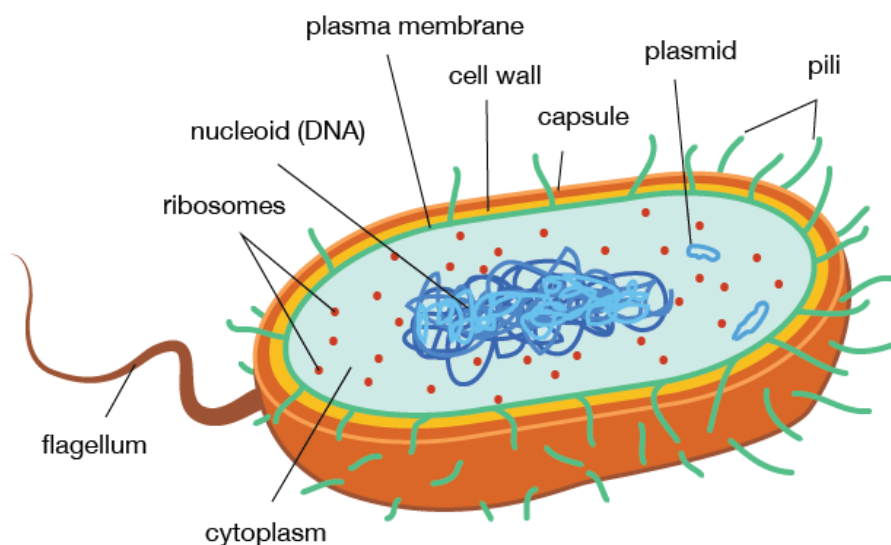


- Golgi Apparatus – a series of fluid-filled, flattened and **curved sacs** with **vesicles** surrounding the edges. The Golgi apparatus **processes and packages proteins** and lipids. It also produces lysosomes.
- Mitochondria – usually oval shaped and bound by a **double membrane** called the envelope. The inner membrane is folded to form projections called **cristae**, with matrix on the inside containing the enzymes needed for cellular **respiration**.
- Centrioles – **hollow cylinders** containing a ring of microtubules arranged at right angles to each other. Centrioles are involved in **cell division**.
- Ribosomes – composed of two subunits. The site of **protein synthesis**.
- Lysosomes – vesicles, containing **digestive enzymes**, bound by a single membrane.

Protein Synthesis and Transport:

- Proteins are **produced on the ribosomes**.
- Proteins which are produced on the ribosomes on the surface on RER are **folded and processed in the RER**.
- The proteins are then **transported from the RER to the Golgi apparatus in vesicles**.
- They are then **modified (e.g. carbohydrates are added to form a glycoprotein) in the Golgi Apparatus**.
- The **Golgi Apparatus packages proteins into vesicles** to be transported around the cells to where they're required. Some of the proteins, such as extracellular enzymes, leave the cell by **exocytosis**.

Ultrastructure of prokaryotic cells, such as bacteria:



- Cell Wall – the cell’s rigid outer covering made of **peptidoglycan**. Provides the cell with **strength and support**.
- Slime Capsule – protective slimy layer which helps the cell to **retain moisture** and **adhere** to surfaces.
- Plasmid – circular piece of DNA.
- Flagellum – a tail-like structure which **rotates to move** the cell.
- Pili – hair-like structures which attach to other bacterial cells.
- Ribosomes – composed of two subunits. The site of **protein synthesis**.
- Mesosomes – **infoldings** of the inner membrane which contain **enzymes required for respiration**.

Mammalian Gametes and Fertilisation

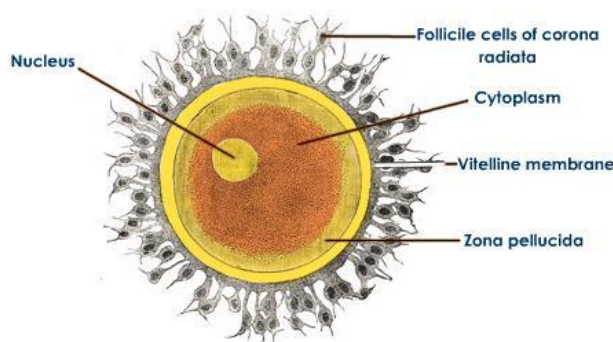


Figure: Tutorvista

Ovum:

- It contains the **zona pellucida** which is a **protective coating** which the sperm have to penetrate in order for fertilisation to occur, the main purpose of zona pellucida is to prevent polyspermy.
- It contains a **haploid nucleus** so that a full set of chromosomes is restored at fertilisation.

- **Cortical granules** release substances which cause the zona pellucida to harden.
- **Follicle cells** form a protective coating around the egg .

Spermatozoa:

- Sperm cell contain many **mitochondria** to provide energy for rotation of the flagellum which enables the cell to move.
- **Acrosome** contains **digestive enzymes** which break down the zona pellucida and allow sperm to penetrate the egg.

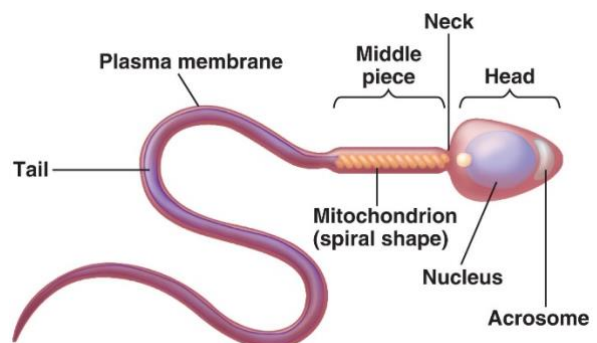


Figure 3 Lifeeasy Biology



Fertilisation:

- 1) The sperm head meets the protective jelly layer around the egg cell called the zona pellucida and the **acrosome reaction** occurs – enzymes digest the zona pellucida.
- 2) The **sperm head fuses with the cell membrane of the egg** cell thus allowing the **sperm nucleus to enter the egg cell**.
- 3) The **cortical reaction** occurs which causes the zona pellucida to harden and prevents polyspermy.
- 4) The nuclei fuse and a **full set of chromosomes** is restored, thus forming a diploid zygote.

The **locus** of a gene is its location on a chromosome. Alleles on the same chromosome are autosomally **linked** and are inherited, to a greater or lesser extent, as if they were the same gene. The closer the loci of the genes on the chromosome, the more closely linked they are. This is because a **recombination event** during meiosis is much less likely to occur between the loci of the two genes.

Some genes are **sex-linked** because they **occur on the X chromosome**. Some genetic disorders are sex-linked and therefore are much more common in men, because they don't have another copy of the allele for a particular characteristic and therefore only need one copy for the allele to be expressed e.g. haemophilia (absence of clotting factor VIII and therefore blood that doesn't clot properly), or red-green colour blindness.

Eukaryotic Cell Cycle and Division

The cell cycle is a process in which cells divide to produce two genetically **identical daughter cells** for growth, repair and asexual reproduction. As all the cells produced by mitosis are genetically identical, mitosis does not give rise to genetic variation.

There are three stages of the cell cycle:

- **Mitosis – prophase, metaphase, anaphase and telophase.**
- **Cytokinesis** – during cytokinesis the **cytoplasm divides**, thus producing two daughter cells.
- **Interphase** – during this stage the cell **grows, DNA replicates and the cell prepares to divide** – chromosomes and some organelles are replicated, chromosomes also begin to condense to form chromatin.

Meiosis is a form of cell division that gives rise to **genetic variation**. The main role of meiosis is **production of haploid gametes and**

Figure: Wikipedia - Gram-negative bacteria



maintenance of chromosome number as cells produced by meiosis have half the number of chromosomes. Meiosis produces genetically different cells, genetic variation is achieved through:

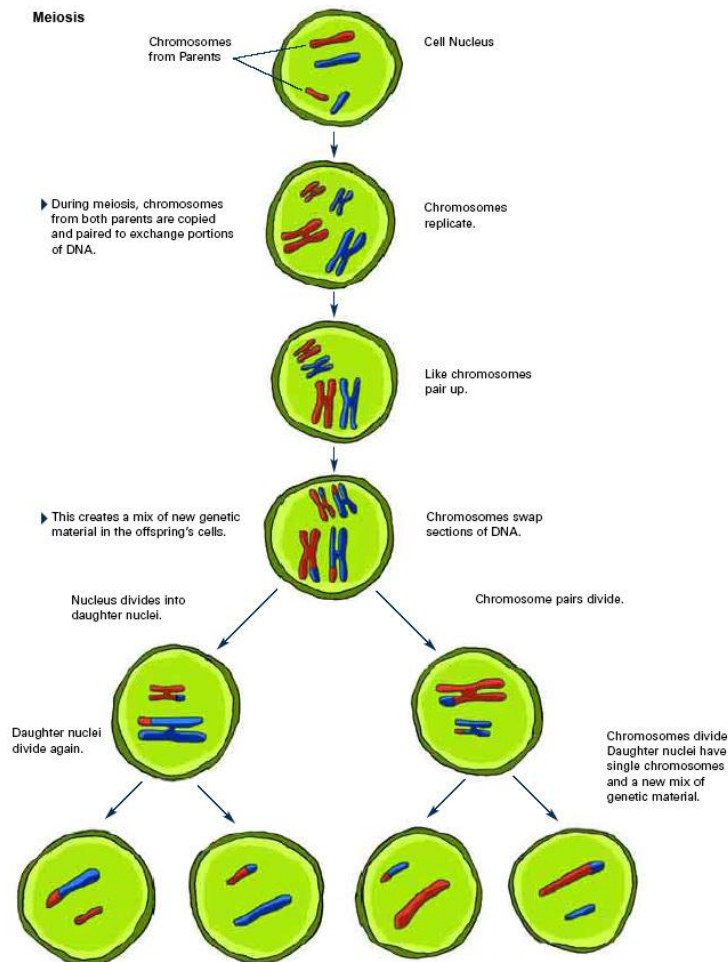


Figure: Biologypost- Meiosis

- **Crossing over** - the exchange of sections of DNA between homologous chromosomes.
- **Independent assortment** – there are various combinations of maternal and paternal chromosome arrangement.

Chromosome mutations can be mutations of the structure of chromosomes (causing **aneuploidy**):

- **Translocation** (swapping of genes, not between alleles in a homologous pair. Involves genes breaking off one chromosome and joining to another). Can be balanced or unbalanced.
- Duplication
- Deletion
- Inversion



Chromosome mutations can also change the number of chromosomes. Aneuploidy happens as a result of **non-disjunction**. This is where either homologous chromosomes or sister chromatids fail to separate. This can result in:

- More than two chromosomes in a pair. This is called **polysomy**.
 - An example of polysomy is Down's Syndrome – trisomy-21
- Less than two chromosomes in a pair, called **monosomy**.
 - An example is Turner's Syndrome, which is monosomy of the sex chromosomes where the one chromosome present is an X.

Stem Cells

Stem cells are undifferentiated cells which can keep dividing to give rise to other cell types. Types of stem cells include:

- **Multipotent** cells, which can give rise to multiple types of cells.
- **Pluripotent** cells which are able to **give rise to many types of specialised cells**, but not placental cells.
- **Totipotent** cells which can **give rise to all types of specialised cells**, including placental cells.

Sources of stem cells include **embryonic stem cells, adult stem cells and fused cells**. Stem cells can be used to **treat a variety of diseases** such as diabetes, multiple sclerosis and Parkinson's disease. They can also be used to **replace damaged tissues** such as nerve tissue in spinal cord injuries. Although there are many **ethical issues** related to the use of stem cells, stem cells **could save many lives and improve the quality of life** of many people. Many people believe use of stem cells is unethical as **embryos are killed in the process** of stem cell extraction. Moreover, there's a **risk of infection when cells are transplanted**, and they could become **cancerous**.

Specialisation of Cells

Differential gene expression allows cells to become specialised and occurs as following:

- A **stimulus** acts on unspecialised cells.
- Some genes become switched on and are **active** as a results whereas other genes are switched off.
- The **active genes are transcribed to produce RNA**.
- **mRNA is then translated** on ribosomes and used to produce protein.
- The protein has the ability **to change the structure and function of cells**.



Phenotype

Phenotype – the **characteristics of an organism**, which result from the **interaction of the genes of the organism with the environment** in which it lives.

There are two types of variation in phenotype – **continuous and discontinuous**. Continuous variation is variation within a range and it includes mass and height whereas discontinuous variation can only take particular values – such as gender or shoe size.

Variation in genotype has an effect on variation in phenotype. Some characteristics are influenced by one gene only and are known as **monogenic**. Such characteristics show **discontinuous variation**. Sometimes times **several genes at different loci** are involved in determining a characteristic – this is known as **polygenic inheritance and often gives rise to continuous variation**.

Some characteristics are influenced by both genotype and the environment. Examples include:

- **Height** is a polygenic characteristic. However, height can be limited due to environmental factors like **nutrition**.
- Some people can be genetically predisposed to **lung cancer** due to the presence of **proto-oncogenes** which regulate the cell cycle. Smoking exposes them to chemicals which convert these genes into active oncogenes in lung cells, thus leading to **uncontrolled cell division** in the lungs, which can result in lung cancer.
- **Animal hair colour**, for example Siamese cat hair colour, is determined by both genotype and environment. Siamese cats have a gene coding for enzyme **tyrosinase** which darkens the fur which is active only below 31 degrees hence **extremities** of Siamese cats are dark.

Epigenetic modifications are modifications to DNA which do not change the base sequence. Epigenetic modifications modify the **activation** of certain genes. Two examples are DNA methylation and histone modification.

DNA methylation is a process by which **methyl groups** are added to DNA. Methylation **modifies the function of the DNA**, typically acting to **suppress gene transcription**. DNA methylation alters the expression of genes in cells as they divide and become specialised. The change is permanent and prevents the cell from converting back into a stem cell or a different cell type.

Histone Modification:

1. **Acetylation** - addition of an acetyl (COCH₃) group- activates chromatin and allows transcription.
2. **Methylation** - addition of a methyl group- can cause activation/inactivation of chromatin depending on the position of the lysine.

