Edexcel (A) Biology A-level

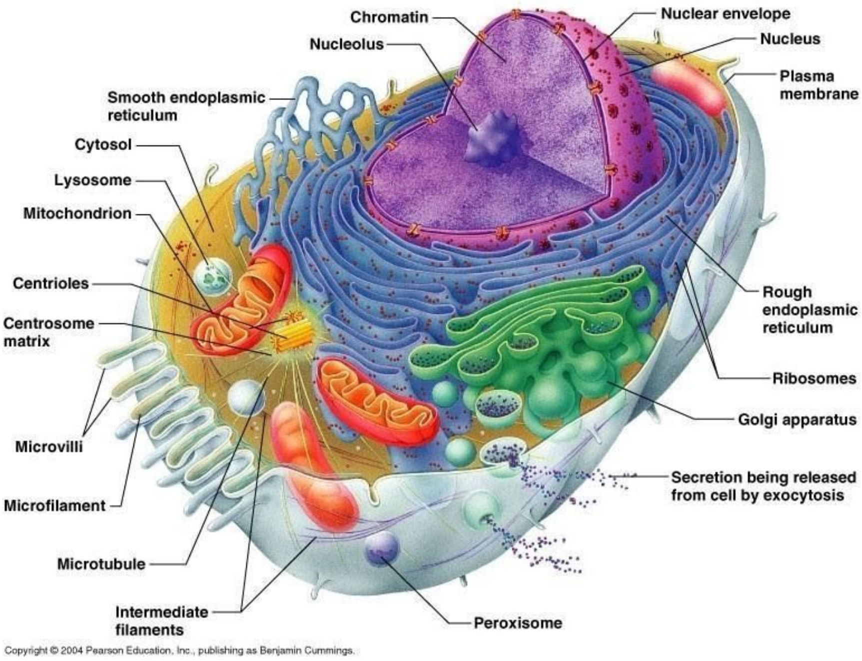
Topic 3: Voice of the Genome

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** - each specialised for a particular function. 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:



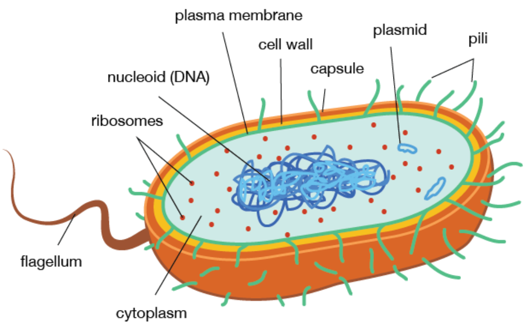
- Nucleus – surrounded by a **double membrane** called the envelope, which contains pores enabling molecules to enter and leave the nucleus. The nucleus contains DNA wrapped around **histone proteins** in a complex called **chromatin**, and a **nucleolus**, which is the site of **ribosome production**.
- 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; often located close to the nucleus.
- 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 **modifies and packages proteins** (after budding off from the RER) as well as 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 a fluid 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.
- 80S Ribosomes – composed of two subunits. The site of protein synthesis.
- Lysosomes – vesicles, containing digestive enzymes, bound by a single membrane.

Protein Trafficking

- 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.

Prokaryotic Cells

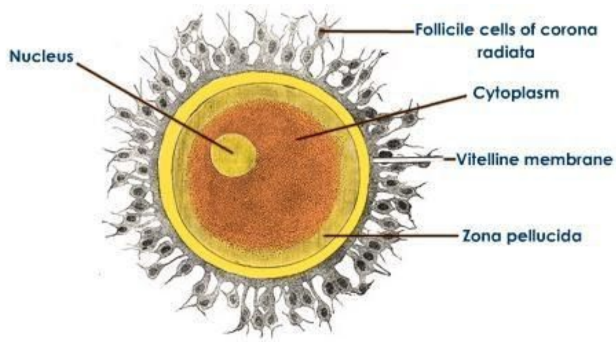


Ultrastructure of prokaryotic cells, such as bacteria:

- 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 pieces of DNA.
- Flagellum – a tail-like structure which rotates to move the cell.

- Pili – hair-like structures which attach to other bacterial cells (allowing the exchange of plasmids).
- 70S Ribosomes – composed of two subunits. The site of **protein synthesis**.
- Mesosomes – **infoldings** of the inner membrane - their function is debated with many scientists believing they are just **artefacts** from the preparation process for microscopy, while others believe they contain **enzymes required for respiration**.

Mammalian Gametes and Fertilisation



Ovum:

- It contains the **zona pellucida** which is a **protective coating** which the sperm have to penetrate in order for fertilisation to occur, it then hardens after they have entered. 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.

Figure: Tutorvista

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

Spermatozoa:

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

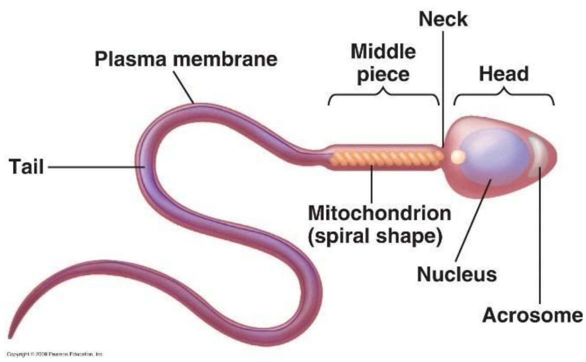


Figure 3 Lifeeasy Biology

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 said to be **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 they are far less likely to be separated during **recombination** in meiosis.

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. This occurs because the Y chromosome is smaller than the X chromosome, so if men don't have another copy of the allele for a particular characteristic. As a result, they 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 colorblindness.

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** – There are three stages within interphase:

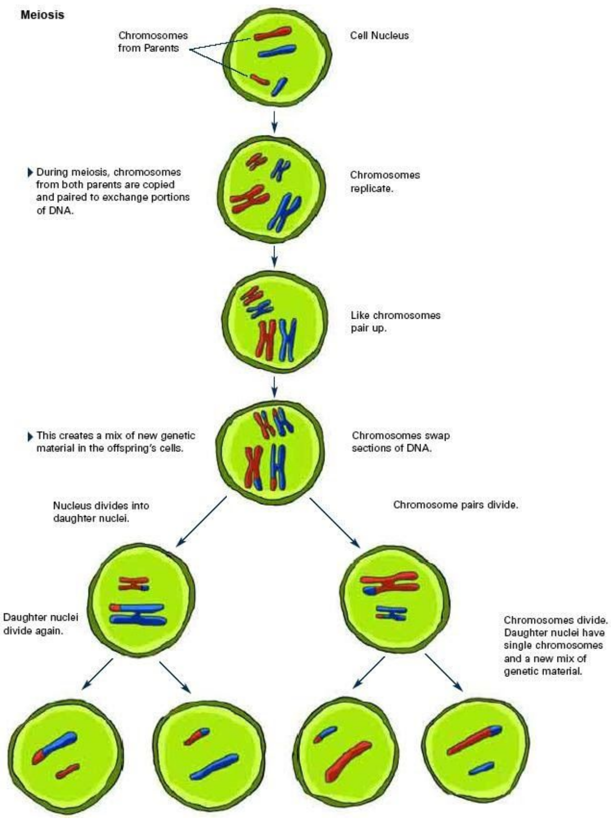
  G1 - during this stage the cell grows

  S – chromosomes are replicated, and begin to condense to form chromatin.

  G2 - the cell prepares to divide, replicating organelles for a full set in each new cell

Meiosis is a form of cell division that gives rise to **genetic variation**. The main role of meiosis is **production of haploid gametes and maintenance of chromosome number** as cells produced by meiosis have half the number of chromosomes (hence it is also known as **reduction division**). Meiosis produces genetically different cells, genetic variation is achieved through:

- **Crossing over** - the exchange of sections of DNA between homologous chromosomes.
- **Independent assortment** – there are various combinations of ways maternal and paternal chromosomes can be distributed between the two daughter cells.



Chromosome mutations can be changes to the structure of chromosomes, or changes to their number (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. There are three types of stem cells:

- Multipotent cells, which can give rise to many different 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 / any types of specialised cells, including placental cells.

Sources of stem cells include embryonic stem cells or adult stem cells (collected from adult bone marrow). 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 embryonic 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.
- Activator and repressor molecules can bind to promoter regions on the DNA sequence. Some genes become switched on and are active whereas other genes are switched off, for example by changing the structure of chromatin, making it more or less open to being freely transcribed. (See below).
- 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 **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**. 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.

Lac Operon

One example of the function of epigenetic changes is the **Lac operon.** This gene in bacteria is suppressed by repressor molecules in the absence of lactose - preventing energy expenditure in transcribing the gene for lactose digesting proteins when lactose is absent. When lactose is present, it prevents the repressor molecule from binding to the operator region of the DNA, allowing the gene to be transcribed, thus its proteins produced.