

## Foreword

Dear student,

Before you lies the booklet for Theme 1a of Semester 1 of Year 1 Medicine at the University of Manchester. Slim Academy has summarized the most important study material for you, so that you can study as pleasantly as possible. We wish you the best of luck with your studies and, of course, with earning your study credits!

### Quality

Because we want to keep the quality of our summaries as high as possible, we are very interested in your feedback. For this reason, we will send you an email two weeks after you receive the summary, asking you to evaluate it. You would really help us by filling it out. If at any other time you have comments, tips, or suggestions for improvement, please email us at **klantenservice@slimacademy.nl** or send us a message via Facebook chat — we'll get to work on it right away!

### Working at Slim Academy

Slim Academy is always looking for motivated students! Would you like to help us by summarizing and reviewing summaries? Then the role of **Study Hero** might be perfect for you. You can work from home, receive a generous payment, and gain a study-related side job that looks great on your CV. Interested? Send your motivation letter and CV to **klantenservice@slimacademy.nl**.

### Reproduction prohibited

Studying together is, of course, effective. So be sure to tell your friends about Slim Academy's summaries. However, reproduction and sharing of this summary with third parties is strictly prohibited. If you want us to continue offering these materials, please don't share this summary with others — encourage your fellow students to get their own copy.

### Get in touch with Slim Academy

Would you like to stay up to date with developments at Slim Academy? Get in touch through:

 [www.slimacademy.nl](http://www.slimacademy.nl)  
 [klantenservice@slimacademy.nl](mailto:klantenservice@slimacademy.nl)  
 @slimAcademy.nl  
 010 214 32 45

We wish you lots of success with your studies and your exams!

### Team Slim Academy

**P.S.** This summary has been written based on the author's own interpretation. It remains a summary and should be seen as a supplement to the required study materials — not a replacement

## Table of content

|  |           |
|--|-----------|
| <b>Foreword</b>  | <b>1</b>  |
| <b>Table of content</b>  | <b>2</b>  |
| <b>Early Embryology, Biology Barriers and an Introduction to Mendelian Inheritance</b>         | <b>3</b>  |
| Chapter 1 - Oogenesis, fertilisation, and cleavage   | 4         |
| Chapter 2 - Major morphogenetic events that occur up to and including gastrulation             | 9         |
| Chapter 3 - Layers of the bilaminar and trilaminar embryo                                      | 11        |
| Chapter 4 - Organelles of a mammalian cell and the differences between male and female gametes | 12        |
| Chapter 5 - Basic types of epithelial cells  | 16        |
| Chapter 6 - Cell signalling  | 18        |
| Chapter 7 - Organisational structure of the skin and the epithelial appendages                 | 21        |
| Chapter 8 - Major functions of the skin  | 23        |
| Chapter 9 - Homeostasis and positive and negative feedback                                     | 24        |
| Chapter 10 - Key terms in genetics   | 25        |
| Chapter 11 - Mendelian inheritance and pedigree diagrams                                       | 26        |
| Chapter 12 - Social and personal impact of living with a long-term health condition            | 31        |
| <b>Afterword</b>   | <b>33</b> |

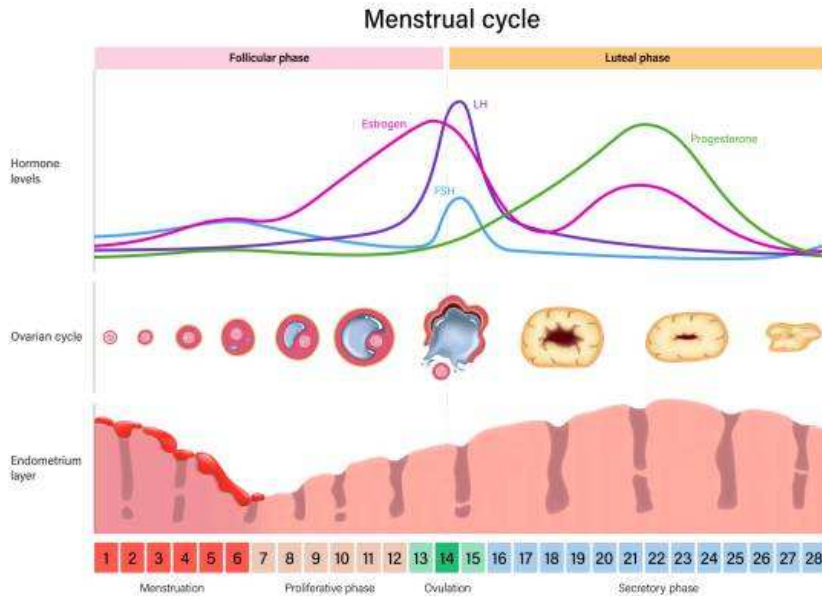
## **Early Embryology, Biology Barriers and an Introduction to Mendelian Inheritance**

**Each chapter is based on the respective ILO. The ILO's can be found on Canvas. Because of copyright protection we will no longer copy the ILO's in this booklet. We recommend you find yourself the ILO's and keep them next to the summary.**

## Chapter 1 - Oogenesis, fertilisation, and cleavage

### Introduction

The chapter details the menstrual and ovarian cycles coordinate and how they contribute to hormonal regulation of the endometrium and ovarian follicles to prepare the female reproductive system for potential fertilisation and pregnancy. This process integrates cyclical changes in estrogen and progesterone with oogenesis, ovulation, fertilisation, and early embryonic development.



**Menstrual cycle.** Source: [www.istockphoto.com](http://www.istockphoto.com)

### Menstrual Phase (Day 1-5)

The menstrual phase is triggered by the decline in progesterone and estrogen following degeneration of the corpus luteum from the previous cycle. The stratum functionalis of the endometrium becomes ischemic as spiral arteries constrict, reducing blood flow and oxygen delivery. This leads to necrosis and shedding of the stratum functionalis, mixed with blood and tissue debris, forming menstrual fluid. The stratum basalis remains intact and serves as the foundation for regeneration. The endometrial lining thins to about 1 to 2 mm with an average blood loss of 30 to 40 mL, and occasionally up to 60 mL. Menstrual blood does not clot because of local fibrinolytic factors that prevent coagulation. This phase coincides with the early development of primary follicles in the ovarian cycle.

### Proliferative or Follicular Phase (Days 6-13)

The primary hormone in this phase is estrogen, secreted by granulosa cells of developing follicles. FSH stimulates follicular growth, and one dominant follicle continues to mature while others undergo atresia. Estrogen secretion from granulosa cells rises and stimulates mitosis of epithelial and stromal cells, leading to regrowth and thickening of the endometrium to about 4 to 10. Estrogen promotes repair and elongation of spiral arteries within the stratum functionalis and the formation of a thin cervical mucus that facilitates sperm entry. Rising estrogen initially exerts negative feedback on FSH and LH, preventing premature ovulation. Toward the end of this phase, sustained high estrogen levels cause a switch to positive feedback, which triggers a surge in LH and FSH, marking the transition to ovulation.

### Ovulation (Day 14)

The LH surge triggers ovulation. When estrogen levels peak, the feedback mechanism becomes positive, causing a rapid increase in LH and a smaller rise in FSH. The LH surge induces completion of meiosis I in the primary oocyte, causes rupture of the follicle and release of the secondary oocyte (arrested in metaphase II) from the Graafian follicle, and promotes luteinization of granulosa and theca cells to form the corpus luteum, which begins secreting progesterone. Ovulation occurs 24 to 36 hours after the onset of the LH surge. The expelled oocyte, surrounded by the zona pellucida and corona radiata, is swept into the ampulla of the uterine tube by the fimbriae.

### **Secretory or Luteal Phase (Days 15–28)**

The primary hormone in this phase is progesterone, secreted by the corpus luteum. Progesterone converts the proliferative endometrium into a secretory one. It stimulates glands to release glycogen, lipids, and glycoproteins to create a nutrient-rich environment for potential implantation. It enhances vascularization through the growth of coiled spiral arteries, inhibits further release of FSH, LH, and GnRH, and exerts immunosuppressive effects to allow for potential embryo implantation. If fertilization does not occur, the corpus luteum degenerates around day 25, forming the corpus albicans. The resulting drop in progesterone and estrogen causes vasospasm of the spiral arteries and ischemic necrosis of the endometrium. The functionalis layer is then shed during menstruation, beginning a new cycle. If fertilization occurs, the developing syncytiotrophoblast of the embryo secretes human chorionic gonadotropin (hCG), which maintains the corpus luteum and progesterone production, sustaining the endometrium until the placenta forms

### **Ovarian Cycle**

Oogenesis is the process by which female gametes (ova) are produced in the ovaries. It begins before birth, pauses during development, and is completed only if fertilization occurs. During fetal life, oogonia undergo mitosis to form primary oocytes. By approximately 12 weeks of gestation, these primary oocytes begin meiosis I but arrest in prophase I. At birth, the ovaries contain about 1–2 million primary oocytes, each surrounded by a single layer of flattened granulosa cells forming a primordial follicle. By puberty, this number decreases to about 300,000. At the onset of puberty, hormonal regulation initiates the menstrual cycle.

### **Oogenesis and Early Follicular Development**

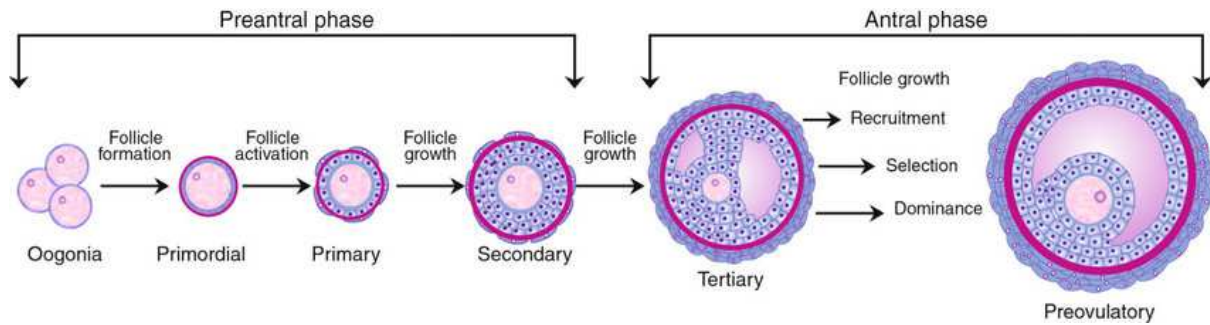
Oogenesis begins before birth and provides the oocytes required for fertilisation and early embryonic development. In the fetal ovary, oogonia undergo mitotic division and differentiate into primary oocytes. Around 12 weeks of gestation, these primary oocytes begin meiosis I but become arrested in prophase I. Each primary oocyte is enclosed within a single layer of flattened granulosa cells, forming a primordial follicle. At birth, the female possesses approximately one to two million of these follicles, which decrease to around 300,000 by puberty.

### **Hormonal Regulation at Puberty**

At the onset of puberty, the hypothalamus begins to secrete gonadotropin-releasing hormone in a pulsatile manner. This stimulates the anterior pituitary gland to release follicle-stimulating hormone and luteinizing hormone. FSH promotes the growth and maturation of ovarian follicles, while LH stimulates theca interna cells to produce androgens, which granulosa cells convert to estrogen via the enzyme aromatase. Rising estrogen levels promote proliferation of granulosa cells and thickening of the endometrium in preparation for possible implantation.

### **Hypothalamic and Pituitary Control**

The hypothalamus regulates follicular development through pulsatile secretion of gonadotropin-releasing hormone. Two nuclei within the hypothalamus, the preoptic and arcuate nuclei, release GnRH into the hypophyseal portal circulation. This hormone acts on gonadotroph cells of the anterior pituitary, stimulating the release of follicle-stimulating hormone and luteinizing hormone. These gonadotropins coordinate follicular growth, steroid hormone synthesis, and oocyte maturation within the ovary



**Follicular Development.** Source: [www.researchgate.net](http://www.researchgate.net)

### Primordial Follicle

The primordial follicle represents the earliest stage of follicular development. Each follicle contains a primary oocyte that is diploid and arrested in prophase I of meiosis. At birth, the ovaries contain approximately one to two million primordial follicles, but by puberty, only about three to four hundred thousand remain. Each primordial follicle is surrounded by a single layer of flattened follicular cells supported by a basal lamina and embedded within the ovarian stroma. The surrounding stromal cells secrete local androgens that help initiate the transition of the primordial follicle into a growing primary follicle. Because the supply of primordial follicles is finite, this pool gradually declines with age.

### Primary Follicle

Primordial follicles begin to grow and become primary follicles. The granulosa cells change from flattened to cuboidal or columnar and start to proliferate around the oocyte. The oocyte remains arrested in prophase I but begins to enlarge and accumulate the cytoplasmic materials required for future divisions, including RNA, lipids, and proteins. A basement membrane separates the growing follicle from the surrounding stroma, maintaining a distinct microenvironment. During this stage, the zona pellucida forms between the oocyte and granulosa cells. This is a glycoprotein-rich extracellular matrix that contains sperm-binding receptors essential for fertilisation.

### Secondary Follicle

As FSH stimulation continues, the primary follicle develops into an early secondary follicle. Granulosa cells proliferate rapidly to form multiple layers around the oocyte. FSH promotes the expression of aromatase in granulosa cells, enabling them to convert androgens into estrogen. At the same time, stromal spindle cells surrounding the follicle differentiate into two layers known as the theca interna and theca externa. The theca interna is highly vascular and contains steroidogenic cells that respond to LH by converting cholesterol into androgens such as androstenedione. These androgens diffuse into the granulosa cells, where FSH-induced aromatase converts them into estradiol. The theca externa is composed of connective tissue and smooth muscle and forms the outer capsule of the developing follicle.

### Antral or Late Secondary Follicle

As the follicle grows, fluid secreted by the granulosa cells begins to accumulate between the cells, forming small fluid-filled spaces. These spaces gradually coalesce to form a central cavity called the antrum. The follicle is now termed a late secondary or early antral follicle. The follicular fluid within the antrum is rich in estrogen, hyaluronic acid, growth factors, and proteins. Continued stimulation by FSH and LH promotes further follicular enlargement and increased estrogen production. The rising concentration of estrogen in the follicular fluid and bloodstream plays an important role in regulating the menstrual cycle through feedback mechanisms.

### **Graafian or Tertiary Follicle**

The Graafian, tertiary, or mature antral follicle represents the final stage of follicular development before ovulation. It contains a secondary oocyte that has completed meiosis I and is arrested in metaphase II. The oocyte is surrounded by the zona pellucida and a crown of granulosa cells known as the corona radiata, which remains attached to the oocyte following ovulation. Within the follicle, the antrum is now large and filled with estrogen-rich follicular fluid. The granulosa cells surrounding the oocyte form the cumulus oophorus, a small mound that anchors the oocyte to the follicular wall. As estrogen production increases, a portion of the hormone is released into the bloodstream and exerts negative feedback on the hypothalamus and anterior pituitary, suppressing further FSH and LH secretion. This mechanism prevents the maturation of additional follicles and allows one follicle to become dominant. Estrogen also acts locally on granulosa cells to enhance their proliferation and increase sensitivity to FSH and LH by upregulating receptor expression. This positive local effect ensures that the dominant follicle continues to grow even as systemic FSH levels decline.

### **Ovulation**

Toward the end of the follicular phase, estrogen levels peak around day fourteen of the menstrual cycle. Sustained high estrogen levels for more than forty-eight hours switch the hypothalamic feedback from negative to positive, resulting in a surge of LH secretion from the anterior pituitary. The LH surge induces enzymatic digestion of the follicular wall, increases vascular permeability, and raises intrafollicular pressure. These changes culminate in rupture of the mature Graafian follicle and release of the secondary oocyte into the peritoneal cavity. The fimbriae of the fallopian tube sweep the oocyte into the ampulla, where fertilisation typically occurs.

### **Luteal Phase and Corpus Luteum**

Following ovulation, the remaining granulosa and theca cells form the corpus luteum, which secretes progesterone and estrogen to maintain the endometrium. If fertilisation does not occur, the corpus luteum degenerates into a corpus albicans, progesterone and estrogen levels fall, and menstruation follows. If fertilisation occurs, the embryo secretes human chorionic gonadotropin which maintains the corpus luteum until the placenta takes over hormone production.

### **Fertilisation**

Fertilisation occurs when a sperm fuses with the secondary oocyte, typically within the ampulla of the fallopian tube. Before fertilisation, sperm must undergo capacitation, a biochemical process triggered by bicarbonate ions in the female reproductive tract that removes surface glycoproteins and cholesterol, increasing sperm motility and membrane fluidity. Capacitated sperm then penetrate the cumulus cells surrounding the oocyte and reach the zona pellucida. Binding to ZP3 receptors on the zona pellucida initiates the acrosomal reaction, during which calcium influx causes fusion of the sperm plasma membrane with the acrosomal membrane, releasing enzymes such as acrosin and proteases that digest a pathway through the zona pellucida.

### **Blocks to Polyspermy and Zygote Formation**

Once a sperm reaches and fuses with the oocyte plasma membrane, two blocks to polyspermy occur. The fast block involves an immediate influx of sodium ions, causing depolarization of the oocyte membrane that temporarily prevents additional sperm from binding. The slow block or cortical reaction follows a rise in intracellular calcium released from the oocyte's smooth endoplasmic reticulum. Calcium triggers exocytosis of cortical granules, which modify and harden the zona pellucida, preventing further sperm penetration. The calcium wave also signals the oocyte to complete meiosis II, producing a mature ovum and a polar body. Fusion of the male and female pronuclei forms a diploid zygote with 46 chromosomes, marking the beginning of a new organism.

### **Cleavage and Early Embryo Transport**

Within 24 hours of fertilisation, the zygote begins mitotic cleavage divisions without an increase in overall size. These successive divisions produce smaller cells called blastomeres, leading to the 2-cell, 4-cell, 8-cell, and 16-cell stages. By the 16-cell stage, the zygote becomes a morula. Compaction occurs as blastomeres tightly adhere to one another through junctional complexes, forming a compact mass surrounded by the zona pellucida. During cleavage, the embryo is transported along the fallopian tube toward the uterine cavity, expressing surface proteins to facilitate implantation.

#### **Slim Summary!**

- Oogenesis is the formation of female gametes in the ovaries and begins before birth. In this process oogonia develop into primary oocytes arrested in prophase I of meiosis. At puberty, FSH stimulates the follicles (primary oocytes) to mature. For every cycle, one primary oocyte completes meiosis I forming a secondary oocyte arrested in metaphase II of meiosis. The process of fertilisation allows the secondary oocyte to complete meiosis II;
- Ovulation consists of the menstrual and ovarian cycle. The menstrual phase (days 1-5) involves the shedding of the endometrial lining due to low oestrogen and progesterone levels after the corpus luteum has degenerated. During the proliferative/follicular phase (days 6-13), FSH stimulates follicle growth while oestrogen from the granulosa cells rebuilds the endometrium lining and makes the cervical mucus thinner. During ovulation (day 14), there is an LH surge which triggers the release of the secondary oocyte from the graafian follicle. Finally in the secretory/luteal phase (days 15-28), the corpus luteum secretes progesterone making the endometrium secretory and vascular. If fertilisation occurs, hCG is secreted which maintains the corpus luteum until the placenta forms, alternatively if there is no fertilisation, the corpus luteum degenerates causing hormone levels to drop and the menstrual cycle restarts;
- Fertilisation occurs in the ampulla of the fallopian tube. Sperm capacitation increases motility and membrane fluidity. The acrosomal reaction releases enzymes to penetrate the zona pellucida. The fusion of the sperm cell and oocyte triggers mechanisms like fast sodium depolarisation and cortical reactions which prevents polyspermy. This fusion also causes the oocyte to complete meiosis II therefore forming a mature ovum and secondary polar body. Finally the male and female pronuclei fuse forming a diploid zygote;
- Cleavage is when the zygote divides mitotically without increasing in size. It starts as 2 cells and doubles until it reaches the morula (16 cell) stage. Compaction occurs as the cells adhere together tightly. Finally the embryo travels along the fallopian tube to the uterus for implantation.

## Chapter 2 - Major morphogenetic events that occur up to and including gastrulation

### Introduction

This chapter introduces blastocyst formation and implantation, differentiation of embryonic tissues, and establishment of the basic body plan through gastrulation and neurulation.

### Blastocyst Formation and Implantation

As the morula enters the uterine cavity around day 5, it begins to absorb uterine fluid, creating a central fluid-filled cavity called the blastocoel. The resulting structure is the blastocyst, consisting of an outer trophoblast layer and an inner cell mass located on one side. The trophoblast will give rise to the placenta, while the inner cell mass will form the embryo. Around days 6 to 7, the blastocyst sheds the zona pellucida and begins implantation into the endometrial lining.

### Trophoblast Differentiation and Decidualization

Implantation occurs between days 7 and 10. The trophoblast differentiates into two layers, an inner cytotrophoblast and an outer syncytiotrophoblast. The syncytiotrophoblast invades the endometrium using proteolytic enzymes that digest extracellular matrix components, allowing the blastocyst to embed. It also secretes human chorionic gonadotropin to maintain the corpus luteum. Small cavities known as lacunae form within the syncytiotrophoblast, which eventually fill with maternal blood as the trophoblast erodes maternal capillaries, establishing early uteroplacental circulation. The endometrium responds to implantation through decidualization, in which stromal cells enlarge and accumulate glycogen and lipids to nourish the developing embryo. Increased vascularization supports nutrient and gas exchange. Chorionic villi begin to form as finger-like projections extending into the lacunae, later developing into secondary and tertiary villi that facilitate maternal-fetal exchange.

### Bilaminar Disc and Early Embryonic Cavities

By the end of the second week, the inner cell mass differentiates into a bilaminar embryonic disc composed of two layers, the epiblast and the hypoblast. The epiblast forms the amniotic cavity above, while the hypoblast contributes to the yolk sac below. Surrounding structures include the amniotic cavity, which cushions the embryo, the primary yolk sac, which is responsible for early nutrient transfer and blood formation, and the chorion, which becomes the principal part of the placenta. The connecting stalk attaches the embryo to the trophoblast and later forms the umbilical cord.

### Gastrulation

Gastrulation begins around day 14 to 16 post-fertilisation and marks the formation of the three germ layers from the bilaminar disc. The process begins with the appearance of the primitive streak along the midline of the epiblast, extending caudally to cranially, with a primitive node at its anterior end. Epiblast cells near the streak undergo an epithelial-to-mesenchymal transition under the influence of fibroblast growth factor 8, which activates the transcription factor SNAIL1, suppressing E-cadherin to allow cell migration. Migrating epiblast cells displace the hypoblast to form the endoderm, then fill the space between epiblast and endoderm to form the mesoderm. Cells remaining in the epiblast become the ectoderm.

### Germ Layers and Their Derivatives

These three germ layers give rise to all tissues and organs of the body. The endoderm forms the epithelial lining of the gastrointestinal and respiratory tracts, as well as associated organs such as the liver, pancreas, thyroid, and urinary bladder. The mesoderm forms muscles, bones, connective tissue, cardiovascular and lymphatic systems, kidneys, and gonads. The ectoderm forms the epidermis, hair, nails, and the entire nervous system.

### **Notochord Formation and Neurulation**

As gastrulation proceeds, the notochord forms from mesodermal cells that migrate through the primitive node. The notochord plays an essential inductive role, stimulating the overlying ectoderm to thicken and form the neural plate. The lateral edges of the neural plate elevate to create neural folds, which converge to form the neural tube, the precursor to the central nervous system. Closure of the neural tube begins in the midregion and progresses both cranially and caudally, with the anterior neuropore closing around day 25 and the posterior neuropore by day 27. Neural crest cells, which detach from the crest of the neural folds, migrate throughout the embryo to form components of the peripheral nervous system, melanocytes, and craniofacial structures.

### **End of the Third Week**

By the end of the third week, gastrulation and neurulation have established the basic body plan of the embryo. The trilaminar germ disc, consisting of ectoderm, mesoderm, and endoderm, forms the foundation from which all organ systems will develop.

#### **Slim Summary!**

- First, the morula develops a fluid filled cavity called a blastocoel, this results in a structure called a blastocyst which contains an outer trophoblast (future placenta) and inner cell mass (future embryo), blastocyst formation occurs around day 5;
- Implantation occurs at around days 6-10, this is where the blastocyst embeds into the endometrium, and the trophoblast differentiates into the cytotrophoblast and syncytiotrophoblast. The syncytiotrophoblast invades the uterine wall and secretes hCG to maintain the corpus luteum while decidualisation of the maternal tissue supports implantation;
- Towards the end of week 2, the bilaminar disc is formed from the inner cell mass as it splits into two layers: the epiblast and the hypoblast. The epiblast forms the amniotic cavity while the hypoblast forms the yolk sac. The chorion also develops as well as the connecting stalk which will become the future umbilical chord;
- The process of forming the trilaminar germ disc from the bilaminar disc is known as gastrulation. During this process, the primitive streak is formed which causes the epiblast cells to migrate to create the endoderm (displaced hypoblast cells), the mesoderm (new layer in between the endoderm and ectoderm), and the ectoderm (the remaining epiblast);
- Notochord formation and Neurulation occurs late week 3, during this process, the mesodermal cells migrating through the primitive node form the notochord which induces the ectoderm to form the neural plate. The neural folds elevate and fuse to create the neural tube while the neural crest cells detach from the neural folds to form the PNS, melanocytes and craniofacial structures;
- By the end of week three the basic body plan is established and the three germ layers of the trilaminar disc give rise to all future tissues and organs.

## Chapter 3 - Layers of the bilaminar and trilaminar embryo

### Introduction

This chapter describes the layers of the bilaminar and trilaminar embryo and their derivatives.

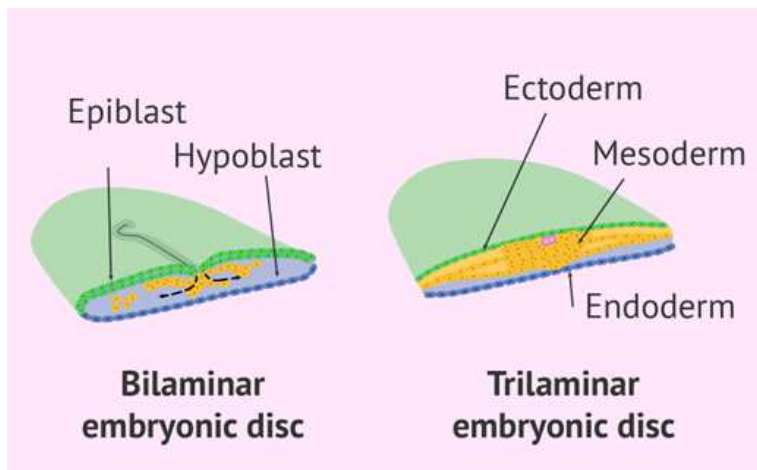
### Bilaminar disc

The bilaminar disc is formed of two layers known as the epiblast and the hypoblast. The **epiblast** is the primitive ectoderm and gives rise to the embryo proper and the three germ layers (ectoderm, mesoderm and endoderm) during gastrulation. The **hypoblast** is the primitive endoderm, which contributes to the extraembryonic membranes including the yolk sac.

During gastrulation, cells from the base of the primitive streak detach and migrate to lie between the layers of ectoderm and endoderm. These cells are invaginated and create the mesoderm. Therefore, a trilaminar disc is formed.

### Trilaminar disc and derivatives

The trilaminar disc is made up of three layers. The first layer is the **ectoderm**, which is the outermost germ layer and forms the nervous system, epidermis, hair and nails. The second layer is the **mesoderm** which is the middle germ layer and forms the musculoskeletal system, circulatory system, and reproductive system. The final layer is the **endoderm** which is the innermost germ layer and forms the gastrointestinal system and the respiratory system.



Layers of the bilaminar and trilaminar disc. Source: invitra.com

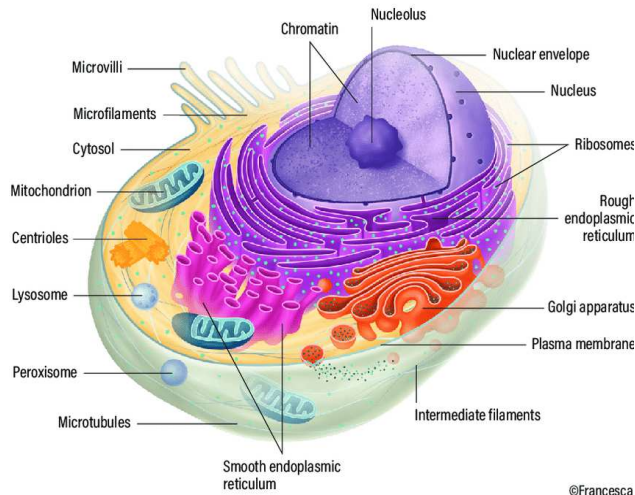
### Slim Summary!

- The bilaminar disc consists of the epiblast and the hypoblast, the epiblast gives rise to the three germ layers of the trilaminar disc;
- During gastrulation, the epiblast cells near the primitive streak migrate creating an extra layer known as the mesoderm - therefore a trilaminar disc is formed;
- The ectoderm is the outermost layer (former epiblast layer), the mesoderm is the middle layer, and the endoderm is the innermost layer (former hypoblast layer).

## Chapter 4 - Organelles of a mammalian cell and the differences between male and female gametes

### Introduction

This chapter outlines the structure and functions of key organelles in a mammalian cell and explains how sperm and ova are specialised to enable fertilisation.



**Structure of a mammalian cell.** Source: [www.researchgate.net](http://www.researchgate.net)

### Nucleus

Contains the cell's chromosomes which are tightly associated with proteins called histones. The nucleus is the site of DNA synthesis, ensuring accurate copying of genetic material during cell division. Transcription occurs here: DNA is used as a template to produce a complementary strand of RNA. Within the nucleus, the nucleolus is responsible for the synthesis and assembly of ribosomal RNA.

### Rough Endoplasmic Reticulum

Function in ribosome production.

### Smooth Endoplasmic Reticulum

Is involved in lipid and steroid synthesis: synthesizes phospholipids, cholesterol, and steroid hormones.

### Golgi Apparatus

Is involved in protein and lipid modification: modifies protein and lipids received from the ER and sorts them into vesicles for delivery to their appropriate destination.

### Plasma Membrane

Selective Permeability: regulates the entry and exit of ions, nutrient, and waste products to maintain cellular homeostasis. It also maintains cellular homeostasis, and interacts with other cells: contains receptor proteins that detect and transmit chemical signals between cells.

### Peroxisome

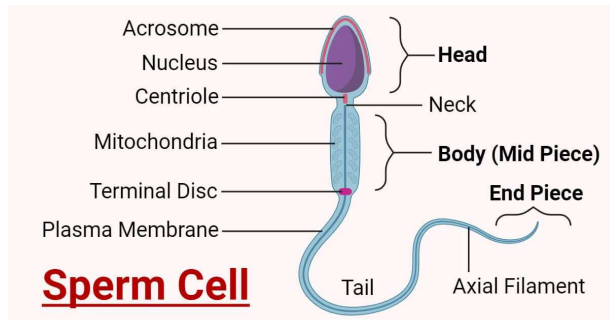
Small membrane bound organelles containing oxidative enzymes. It is involved in lipid metabolism: breakdown of fats and detoxification: detoxifies alcohol and other harmful substances in the liver.

### Lysosome

Membrane-bound vesicles containing hydrolytic enzymes. It carries out intracellular digestion: breaks down unwanted materials such as damaged organelles, ingested bacteria, and macromolecules. Furthermore, lysosomes can carry out apoptosis: Can release enzymes that initiate programmed cell death when necessary.

### Mitochondria

Involved in ATP production: Site of cellular respiration, generating ATP through oxidative phosphorylation. It's semi-autonomous: contains its own DNA and ribosomes, it can synthesize some of its own proteins.



**Structure of Sperm.** Source: <https://microbes.com>

### Acrosome

Is a cap-like structure on the head of a sperm. It contains acrosomal enzymes such as hyaluronidase and acrosin that helps the sperm penetrate the zona pellucida during fertilisation.

### Sperm Centrioles

Are found in the neck region of sperm that separates the head and tail. It is involved in sperm Development: facilitates mitotic and meiotic divisions during sperm development. It also acts as a structural link between the head and tail of the sperm. Furthermore it enables the sperms whip-like motion necessary for motility. Post fertilisation sperm centrioles help in organising and shaping the zygotes cytoskeleton.

### Axoneme

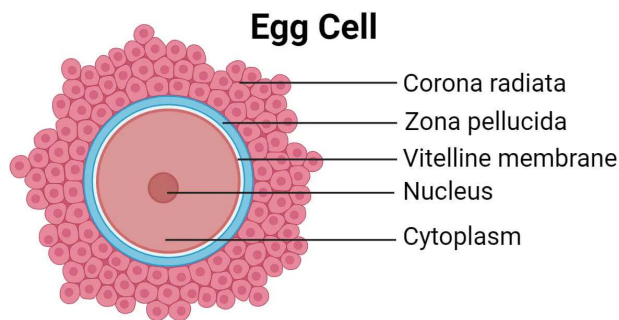
Core structural component of the sperm tail (flagellum), responsible for generating movement. The axoneme contains a specific arrangement of microtubules — nine peripheral doublets surrounding two central singlet microtubules. The movement of the axoneme is powered by dynein arms, motor proteins that cause sliding of adjacent microtubule doublets, producing the whip-like motion of the tail. Regional Organization of the Axoneme: Middle Piece: surrounded by a mitochondrial sheath, where mitochondria generate ATP to fuel dynein activity, Principal Piece: enclosed by a fibrous sheath, which provides mechanical support, End Piece: This terminal region lacks the fibrous sheath, facilitating increased flexibility for fine control of motion.

### Terminal Disc

Links the flagella to the axoneme.

### Flagella

Coordinated wave-like beating of the flagellum propels the sperm forward, enabling motility.



**Structure of the Ovum (Egg Cell).** Source: <https://microbenotes.com>

### Corona Radiata

**Structure:** Multiple layers of granulosa cells that directly surround the zona pellucida. **Origin:** Derived from the cumulus oophorus, which supports the oocyte during follicular development. **Connections:** Corona radiata cells are connected to the oocyte plasma membrane (oolemma) via microvilli and transzonal projections forming gap junctions. **Function:** Provides metabolic and nutritional support to the oocyte, Facilitates the transfer of ions, amino acids, and signaling molecules, and Aids in sperm guidance toward the oocyte during fertilization, it also offers a protective cushion during ovulation and transport in the fallopian tube.

### Zona Pellucida

**Structure:** A thick, transparent, glycoprotein-rich extracellular matrix surrounding the plasma membrane of the oocyte. **Composition:** Composed mainly of glycoproteins ZP1, ZP2, and ZP3. ZP3 functions as the primary sperm receptor, binding to complementary molecules on the sperm membrane to initiate the acrosomal reaction. ZP2 facilitates the binding and passage of acrosome-reacted sperm through the zona. **Function:** Provides mechanical protection to the oocyte and early embryo, mediates species-specific sperm recognition and binding, after fertilization, undergoes biochemical modification (the zona reaction) to prevent polyspermy by inactivating sperm-binding receptors (mainly ZP3).

### Plasma Membrane (Oolemma)

**Structure:** A selectively permeable lipid bilayer that encloses the oocyte cytoplasm. **Function:** Regulates exchange of ions and molecules between the oocyte and its surroundings, contains sperm-binding receptors essential for gamete fusion, triggers the cortical reaction after sperm fusion to prevent polyspermy.

### Cortical Granules

**Structure:** Membrane-bound vesicles located beneath the oolemma in the oocyte cortex. **Function:** upon sperm entry, these vesicles undergo exocytosis, releasing enzymes and polysaccharides into the perivitelline space, the released enzymes modify the zona pellucida glycoproteins (especially ZP3), hardening it and preventing additional sperm from binding or penetrating (slow block to polyspermy). This process ensures that only one sperm fertilizes the ovum.

### Nucleus and Cytoplasm (Ooplasm)

The nucleus contains the haploid maternal genome and is arrested at metaphase II until fertilization. Upon sperm entry, meiosis II completes, producing the mature ovum and a second polar body. The cytoplasm is rich in mRNA, ribosomes, proteins, and nutrients essential for early embryonic development before implantation. After fertilization, the female pronucleus fuses with the male pronucleus to form the diploid zygote.

### **Perivitelline Space**

Location: Narrow space between the oocyte plasma membrane and the zona pellucida. Contains the first and second polar bodies following meiotic divisions. During fertilization, cortical granule contents are released into this space to modify the zona pellucida, contributing to the block to polyspermy.

#### **Slim Summary!**

- The main organelles of a mammalian cell consists of: the nucleus (carries the genetic material), rough ER (site of protein synthesis), smooth ER (lipid and steroid synthesis), golgi apparatus (modifies, sorts, and packages proteins/lipids into vesicles), plasma membrane (selective permeability), peroxisomes (lipid metabolism), lysosomes (intracellular digestion), and mitochondria (ATP production);
- The specific organelles found in the male gametes (sperm) includes: the acrosome (contains enzymes to penetrate through the zona pellucida), centrioles (organise the cytoskeleton), axoneme (split into the middle piece, principal piece, and the end piece), and the flagellum (important for motility);
- The specific organelles found in the female gametes (ovum) includes: the corona radiata (provides nutrients, metabolic support, and sperm guidance), zona pellucida (blocks polyspermy), plasma membrane/oolemma (selective barrier), cortical granules (hardens the zona pellucida), nucleus (haploid genome), cytoplasm/ooplasm (rich in nutrients for early development), and perivitelline space (contains polar bodies).

## Chapter 5 - Basic types of epithelial cells

### Introduction

This chapter describes the basic types of epithelial cells and explains how their structure is modified for their function.

### Simple squamous

Simple means that the epithelium is only one cell thick and squamous means that the cells are flat. Its function is to exchange nutrients and gases and it is found in blood vessels, chambers of the heart, and alveoli.

### Non-keratinised stratified squamous

This type of epithelium is flat, as it is squamous. It is stratified, which means that it consists of several layers. Its function is to behave as a protective barrier, and it's found in the oral cavity, anus, vagina, oesophagus, and skin.

### Keratinised stratified squamous

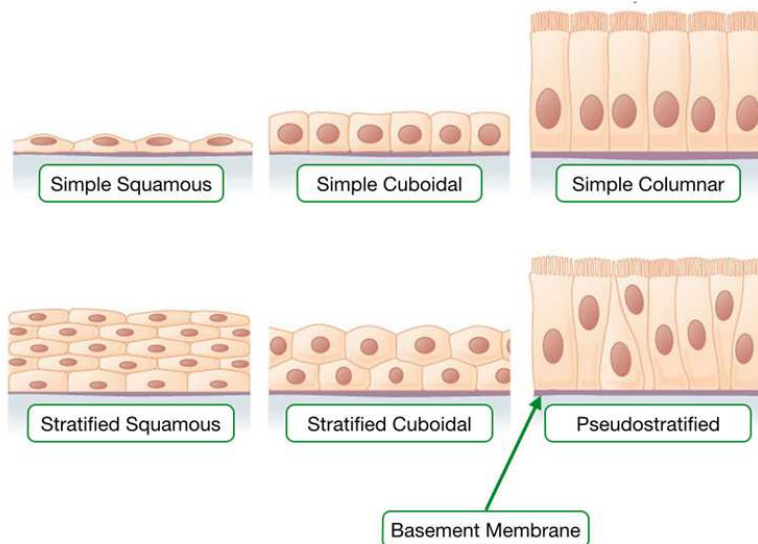
This type of epithelium is also flat and consists of several layers consisting of a keratin layer and a granular layer. It acts as a protective waterproof barrier and is found in the epidermis.

### Cuboidal

This type of epithelium contains cube-shaped epithelial cells where the height is approximately equal to the width. Its functions are involved in absorption and secretion, and it is found in the salivary glands, kidney tubules, and pancreas.

### Simple columnar

Columnar cells are tall with basally located nuclei and, as it is simple epithelium, it is one cell thick. Its functions include absorption and secretion. It is found in the gastrointestinal tract, such as in the small intestine. The cells may also be modified with structures specific to function, for example cilia, microvilli or stereocilia.



Basic types of epithelial cells. Source: [ar.inspiredpencil.com](http://ar.inspiredpencil.com)

### **Specialised epithelia: Pseudostratified columnar epithelium with goblet cells**

This epithelium appears to be stratified with several layers of cells, but there is only one layer of cells. This is because there are many cells crowded together, but all the cells still touch the basement membrane. They contain goblet cells that secrete mucus. It behaves as a mucociliary escalator and is found in the trachea and large respiratory airways.

### **Specialised epithelia: Transitional epithelium**

Transitional epithelium is found in the bladder. The epithelial cells lining the bladder change their shape to allow filling. If the bladder is empty, the shape is cuboidal, and if the bladder is full the cells are squamous.

#### **Slim Summary!**

- Epithelial tissue covers the body surfaces, lines cavities, and forms glands. Their cell shape, layers, and specialisations allow them to perform specific roles including protection, absorption, secretion, and exchange;
- Epithelial tissue can be classified according to the shape of the individual cells and how the cells are arranged;
- Epithelial cells can either be: squamous (squashed), columnar (taller than they are wide), or cuboidal (equal width and height). These cells can be arranged in layers (stratified epithelium), or can be one cell thick/a single layer (simple epithelium);
- Epithelial cells can be modified to have specialised adaptations, e.g. the presence of keratin, cilia, microvilli, or goblet cells;
- pseudostratified epithelium is a specialised type of epithelium which appears to look multi-layered however all the cells touch the basement membrane;
- Transitional epithelium is another type of specialised epithelium only found in the bladder, the epithelial cells that make up this tissue change shape depending on whether the bladder is empty or full.

## Chapter 6 - Cell signalling

### Introduction

This chapter describes the basic elements of cell signalling from external cues to nuclear events that maintain normal cell proliferation and migration. Signals discriminate by receptor compatibility and the response to a ligand changes over time.

### Steps

The first event of signalling is the biosynthesis and release of a signal. Next, dissemination of the signals to the target cell occurs. Signal detection & transduction then takes place which leads to alteration of the cell phenotype. Finally, the signal is terminated.

### Autocrine Signalling

In autocrine signalling, the cell targets itself. This is crucial during human development as it helps cells to assume correct identities and reinforces roles. For example, morphogens are signals that form gradients and cause different developmental effects based on concentration. Shh is an example of a morphogen. Autocrine signalling is also involved in cancer and metastasis.

### Paracrine Signalling

This is where cells target a nearby cell. This type of signaling is also crucial during development as it enables one group of cells to instruct a neighbouring group on its cellular identity. It is locally coordinating activities with neighbouring cells. Synapses are very fast paracrine systems.

### Endocrine and Exocrine Signalling

**Endocrine** signalling uses the circulatory system to transmit signals over long distances. Specialised cells release hormones into the bloodstream to target cells, then glands release hormones that act as master regulators of the development of physiology. Examples of glands include the thyroid, hypothalamus, pituitary, and pancreas. **Exocrine** signalling involves releasing signals into ducts instead of the bloodstream.

### Ligand-gated ion channels

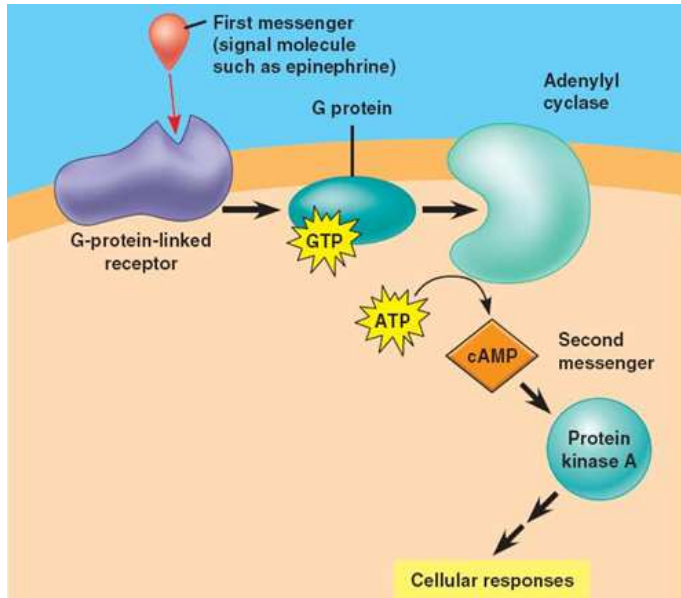
These ion channels are transmembrane proteins that open in response to a ligand binding. This binding allows specific ions to flow across the membrane, which alters membrane potential and so facilitates responses. Examples include nicotinic receptors and GABA receptors. These channels may have multiple functions depending on receptor type.

### Cell-cell connections - Gap Junctions

This involves direct communication between adjacent cells. Cells are held together by interlocking membrane proteins containing a central pore. This allows the direct passage of ions and molecules between cells, which enables fast, co-ordinated responses. These connections are found in cardiac and smooth muscle.

### G-Protein Coupled Receptors (GPCRs)

G-Protein coupled receptors are a large family of receptors that detect signals. G-proteins are activated when ligands bind, which initiates intracellular responses. Binding can lead to changes in enzyme activity e.g. adenylate cyclase or the production of second messengers e.g. cAMP, IP<sub>3</sub>. These receptors are common drug targets.



**cAMP as a second messenger.** Source: [bio1151b.nicerweb.com](http://bio1151b.nicerweb.com)

### Receptor Tyrosine Kinases

These are cell surface receptors that phosphorylate tyrosine residues on themselves and other proteins in response to ligand binding. They are activated by various ligands. Key pathways include MAPK and PI3K/Akt pathways. Dysregulation of RTKs is implicated in cancers, making them important therapeutic targets.

### Hydrophilic and Hydrophobic ligands

**Hydrophilic** ligands are water-soluble, so cannot cross the phospholipid bilayer. They bind to extracellular receptors and trigger signal transduction via secondary messengers. Peptide hormones are hydrophilic. **Hydrophobic** ligands are lipid-soluble, so they can diffuse across the phospholipid bilayer of the cell membrane, bind to intracellular receptors, and often regulate gene expression. Steroid hormones are hydrophobic.

### Phosphorylation (regulator)

This involves the addition of a phosphate group to protein and other molecules. It affects enzymes, signal transduction, and protein interactions. Phosphorylation is catalysed by kinases and reversed by phosphatases.

### Phospholipid signalling

This involves the breakdown of membrane phospholipids by phospholipases e.g. phospholipase C.  $PIP_2$  is hydrolysed into inositol triphosphate, which is a second messenger that triggers calcium ion release. DAG activates protein kinase C which leads to cellular responses. This process is crucial in cell growth, differentiation and immunity.

### Slim Summary!

- Cell signalling is important as it allows cells to efficiently communicate with each other and coordinate their activities. Cells communicate using chemical signals/ligands which are produced by a sending cell, target cells then detect these signals as they have the right receptor for that specific ligand;
- There are a variety of different types of signalling which can be distinguished based on the distance the signal travels before it reaches its target. Paracrine signalling is when cells communicate over short distances, autocrine signalling is when a cell sends a signal to itself, endocrine signalling is when cells transmit signals over long distances;
- Cell signalling receptors can be either found inside the cell (intracellular receptors), or found in the plasma membrane;
- The binding of a ligand to a receptor triggers a signalling pathway which eventually leads to a response.

## Chapter 7 - Organisational structure of the skin and the epithelial appendages

### Introduction

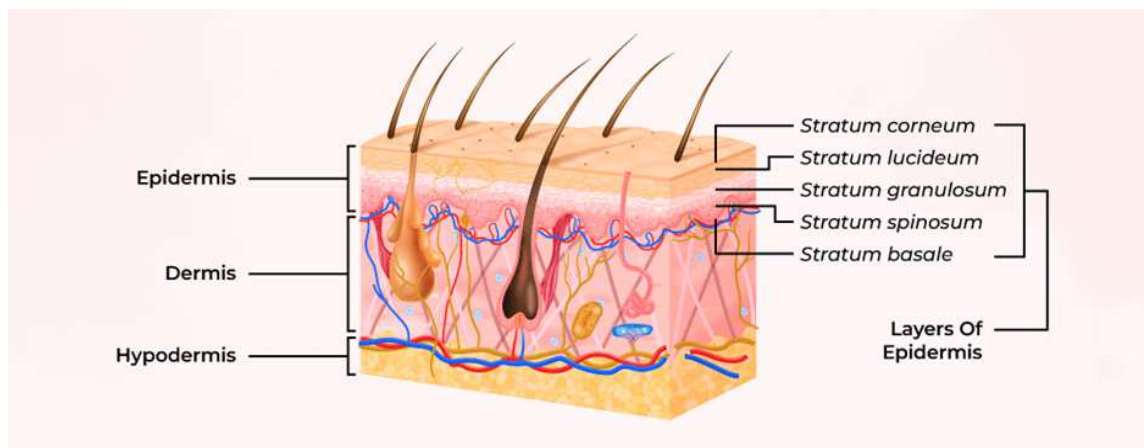
This chapter describes the organisational structure of the skin and the epithelial appendages.

The **epidermis** is made up of multiple layers. The bottom layer is the **stratum basale**, a single layer of cells where mitosis occurs. There are also melanocytes in the stratum basale, which produce melanin that protects against UV radiation. The **stratum spinosum** is a prickle cell layer, which includes 8-10 layers of cells characterised by spines or cytoplasmic processes that connect neighbouring cells. This layer provides strength and flexibility. The **stratum granulosum** consists of 3-5 layers of cells containing granules that are precursors to keratin. The **stratum lucidum** is a thin, transparent layer of cells which is only in thick areas of skin, such as the palms of hands and the soles of the feet. It consists of keratinocytes that have accumulated keratin. Finally, the top layer of keratinised stratified squamous epithelium is termed the **stratum corneum**.

The **dermis** consists of dense irregular connective tissue that resists stretching and distension. It contains: bundles of collagen fibres, elastic fibres, little ground substance and fibroblasts, blood vessels and nerves throughout. The dermis is subdivided into the papillary layer (outer portion), and the reticular layer.

The **subcutaneous layer**, also known as the **hypodermis** or **superficial fascia** is areolar connective tissue that binds the skin tightly to organs. It contains varying amounts of adipose tissue, some collagen fibres, and has an extensive vascular supply.

Epithelial appendages include the hair follicles and hairs, which extend from the subcutaneous layer. The base is expanded at the hair bulb which encloses the hair papilla. These hairs are involved in regulating temperature. The sebaceous glands are found in the dermis and they produce sebum which is released by holocrine secretion. Sebum is a lipid that maintains hair and has some weak anti-bacterial and anti-fungal properties.



Layers of the skin. Source: [geeksforgEEKS.org](https://www.geeksforgEEKS.org)

### Slim Summary!

- The skin is made up of three main layers. The surface layer is known as the epidermis, the mid layer is known as the dermis/dermal layer, the third and deepest layer is known as the hypodermis;
- The epidermis is divided into many sublayers, starting from the base the layers are: basalis, spinosum, granulosum, lucidum, corneum;
- The dermis is found between the epidermis and the hypodermis, this layer is subdivided into the papillary layer (outer portion of the dermis) and the reticular layer (the rest of the dermal layer);
- The hypodermis is found beneath the dermis and is made up of adipose tissue, collagen fibres, and large blood vessels.

## Chapter 8 - Major functions of the skin

### Introduction

This chapter details the major functions of the skin.

The skin has several functions, one of these is protection from harmful microorganisms as it forms a physical barrier. It also acts as a sensory organ, containing several types of sensory receptors that detect touch, pain, pressure, and temperature.

The skin also has hairs which aid in temperature regulation. These hairs extend from the subcutaneous layer and have an expanded base known as the hair bulb which encloses the hair papilla. The hair papilla is a structure of connective tissue which supplies the hair with nutrients essential for its growth. When the body is too cold, the arrector pili muscles contract, and the hairs stand up to trap an insulating layer of warm air. There are sebaceous glands in the dermis, which produce sebum released by holocrine secretion. Sebum is a lipid that maintains the hair and has some weak anti-bacterial and anti-fungal properties.

Melanocytes in the stratum basale produce melanin which protects against UV radiation. There are also macrophages known as Langerhans cells in the epidermis, which initiate immune responses.

### Slim Summary!

- The skin is specialised to carry out a variety of functions. It serves as protection as it forms a physical barrier against microorganisms;
- It helps to regulate body temperature through the presence of sweat glands and hair follicles;
- It contains sensory receptors that detect pain, temperature, touch, and pressure;
- The skin also protects against UV radiation and is involved in wound healing.

## Chapter 9 - Homeostasis and positive and negative feedback

### Introduction

Homeostasis is the process by which the body maintains a stable internal environment despite changes in the external or internal conditions. It involves continuous monitoring and regulation of variables such as body temperature, blood pressure, pH, and glucose levels within narrow limits that are optimal for cellular function.

Negative Feedback: Reverses a change to keep variables near a set point.

Key Examples:

- Blood glucose: High glucose → pancreas releases insulin → cells take up glucose → levels fall → insulin secretion stops.
- Body temperature: High temp → hypothalamus triggers sweating/vasodilation → temp falls.
- Blood pressure: Low BP → baroreceptors activate sympathetic response → HR and vasoconstriction increase → BP normalizes.

Positive Feedback: Amplifies a change until a specific endpoint is reached.

Key Examples:

- Childbirth: Cervical stretch → oxytocin release → stronger contractions → more stretch → continues until birth.
- Blood clotting: Platelet activation → more platelet recruitment → clot forms → loop ends when vessel sealed.

### Slim Summary!

- Homeostasis is defined as the process by which the body maintains a relatively stable internal environment (within a narrow range) despite changes in internal or external conditions/environment. This control is achieved by different physiological processes and is important as it ensures normal cell functioning;
- Negative feedback reverses a change to bring conditions back to normal and decreases the effect of the original stimulus, examples of negative feedback include blood pressure, temperature, blood glucose levels. While positive feedback amplifies a change until certain conditions are met and increases the effect of the original stimulus, examples of positive feedback include blood clotting, and the effects of oxytocin and labour.

## Chapter 10 - Key terms in genetics

### Introduction

This chapter defines the key terms in genetics

- **Genotype:** The genetic constitution of an individual, referring to the specific alleles present at one or more loci.
- **Phenotype:** The observable physical or biochemical characteristics of an organism, resulting from the interaction of its genotype with the environment.
- **Heterozygous:** Having two different alleles at a particular gene locus.
- **Hemizygous:** Having only one allele for a gene instead of the usual two, often used in reference to genes on the X chromosome in males.
- **Congenital:** Referring to a condition that is present from birth, which may be inherited or caused by environmental factors during pregnancy.
- **Dominant:** An allele that expresses its phenotype even when only one copy is present in the genotype.
- **Recessive:** An allele that only expresses its phenotype when two copies are present in the genotype.
- **Allele:** A variant form of a gene that can result in different traits; individuals inherit one allele from each parent.
- **Autosome:** Any chromosome that is not a sex chromosome; humans have 22 pairs of autosomes.
- **Sex Chromosome:** Chromosomes that determine the sex of an individual, typically the X and Y chromosomes in mammals.

### Slim Summary!

- Understanding these genetic terms is important as they form the foundations of genes and inheritance;
- The genotype refers to all the alleles an individual carries. Phenotype is the observable characteristics of an individual. Heterozygous means having two different alleles for a particular gene. Hemizygous refers to only having one allele of a gene. Congenital refers to a condition that is present from birth. A dominant allele is expressed even when only one copy is present. A recessive allele is only expressed when there are two copies present. Alleles are different forms of a gene. Sex chromosomes are chromosomes that determine the sex of an individual, in humans it includes XX (female) and XY (male).

## Chapter 11 - Mendelian inheritance and pedigree diagrams

### Introduction

This chapter explains the main forms of Mendelian inheritance and gives examples of them in pedigree diagrams.

### Key concepts

Patterns of inheritance can involve dominant or recessive genes that cause disease. For every gene, each parent contributes a randomly selected allele to their offspring. This is defined as segregation. Separate genes are independently assorted, which means that separate genes for separate traits are passed to offspring independently unless the genes are linked. If genes are linked (on the same chromosome) then they are sorted as a unit. Note that when there is a percentage risk of offspring having the condition, the chance is the same for each child and does not increase or decrease for future offspring. Therefore, if there is, for example, a 50% chance of offspring having a condition, it does not necessarily mean that exactly half of the children will have the condition. **Pedigree diagrams** visualise how traits are inherited in families. Circles are used to represent females, and squares are used to represent males. When an individual is affected, they appear as shaded in the diagram.

### Autosomal dominant

In autosomal dominant inheritance, only one copy of the faulty allele is required to present with the condition.

For example, if one parent has an autosomal dominant condition:

Parent Genotypes: Aa aa

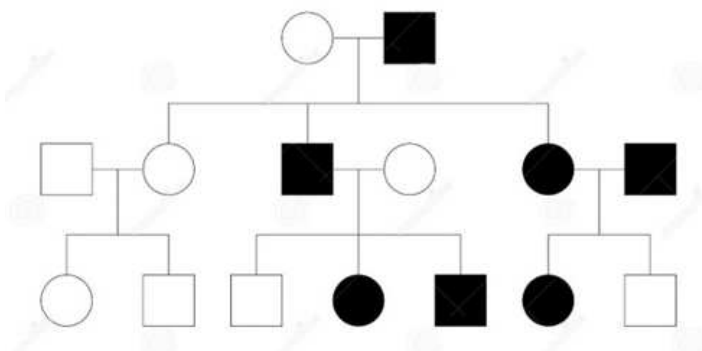
Gametes: A a a a

Possible genotypes: Aa, aa (1:1)

|   | A  | a  |
|---|----|----|
| a | Aa | aa |
| a | Aa | aa |

**Autosomal dominant punnett square.** Source: SlimAcademy

There is a 50% chance of each child having the condition.



**Autosomal dominant pedigree diagram.** Source: dreamstime.com

In an autosomal dominant pedigree, all affected children will have an affected parent. Males and females should be equally affected.

Examples of autosomal dominant conditions include autosomal dominant polycystic kidney disease and Huntington's disease.

**Autosomal recessive**

In autosomal recessive inheritance, two copies of the faulty allele are required for the condition to present. Carriers will have one copy of the faulty allele but won't present with the condition.

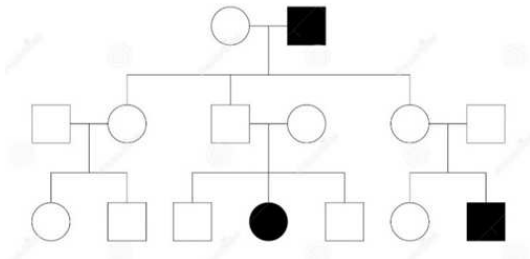
For example, if both parents are carriers of the recessive faulty allele:

- Parent genotypes: Bb Bb
- Gametes: B b B b
- Possible genotypes: BB, Bb, bb (1:2:1)

|          |          |          |
|----------|----------|----------|
|          | <b>B</b> | <b>b</b> |
| <b>B</b> | BB       | Bb       |
| <b>b</b> | Bb       | bb       |

**Autosomal recessive punnett square.** Source: SlimAcademy

There is a 25% chance for each child of having the condition.



**Autosomal recessive pedigree diagram.** Source: dreamstime.com

In an autosomal recessive pedigree, there are affected children with unaffected parents. Males and females should be equally affected.

Cystic fibrosis is an example of an autosomal recessive condition.

**X-linked dominant**

In X-linked dominant inheritance, only one affected X chromosome is required to present with the condition.

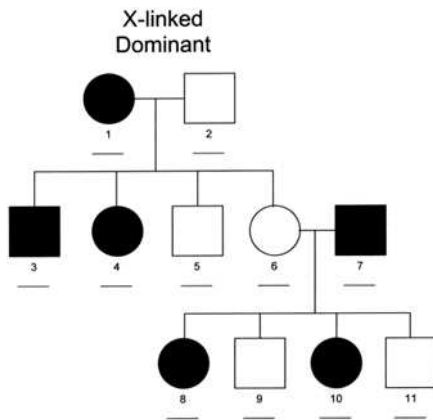
For example, if the father has the condition:

- Parent genotypes: X<sup>R</sup>Y X<sup>r</sup>X<sup>r</sup>
- Gametes: X<sup>R</sup> Y X<sup>r</sup> X<sup>r</sup>
- Possible genotypes: X<sup>R</sup>X<sup>r</sup>, X<sup>r</sup>Y (1:1)

|                      |                               |                  |
|----------------------|-------------------------------|------------------|
|                      | <b>X<sup>R</sup></b>          | <b>Y</b>         |
| <b>X<sup>r</sup></b> | X <sup>R</sup> X <sup>r</sup> | X <sup>r</sup> Y |
| <b>X<sup>r</sup></b> | X <sup>R</sup> X <sup>r</sup> | X <sup>r</sup> Y |

**X-linked dominant punnett square.** Source: SlimAcademy

All daughters will have the condition, whilst no sons will have the condition. Therefore, there is no male-male transmission, because a son will always inherit the Y chromosome from his father, not the affected X chromosome. This is shown in individual 6 and 7's offspring in the pedigree diagram below, where 7 is an affected father.



**X-linked dominant pedigree diagram.** Source: *migr.org*

In an X-linked dominant pedigree diagram, generally more females are affected than males. As mentioned before, there is also no male-to-male transmission because sons always inherit the Y chromosome from their father. If a male is affected, it is because they inherited the affected X chromosome from their mother.

Examples of X-linked dominant conditions are Rett syndrome and fragile X syndrome.

**X-linked recessive**

In X-linked recessive inheritance, women require two affected X chromosomes to present with the condition, whilst men only require one affected X chromosome as they do not have a second X chromosome to compensate. Women with one affected X chromosome are carriers of the condition but do not present with it.

For example, if the father has the condition:

Parent genotypes:  $X^BY$   $X^GX^G$

|       |          |        |
|-------|----------|--------|
|       | $X^G$    | $Y$    |
| $X^G$ | $X^GX^G$ | $X^GY$ |
| $X^G$ | $X^GX^G$ | $X^GY$ |

**X-linked recessive punnett square.** Source: *SlimAcademy*

There is a 0% chance of offspring presenting with the condition, but all the daughters will be carriers. Like in X-linked dominant inheritance, there is no male-to-male transmission as sons always inherit the Y chromosome from their father and not the affected X chromosome.

Then, if one of the carrier daughters has offspring:

Parent genotypes:  $X^GY$   $X^GX^g$

Gametes:  $X^G$   $Y$   $X^G$   $X^g$

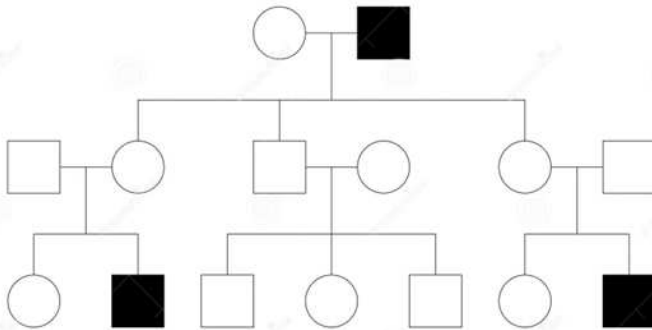
Possible genotypes:  $X^GX^g$ ,  $X^GX^G$ ,  $X^GY$ ,  $X^gY$  (1:1:1:1)

|       |          |        |
|-------|----------|--------|
|       | $X^G$    | $Y$    |
| $X^G$ | $X^GX^G$ | $X^GY$ |
| $X^g$ | $X^GX^g$ | $X^gY$ |

**X-linked recessive punnett square.** Source: *SlimAcademy*

Sons will have a 50% chance of having the condition. No daughters will present with the condition, but some may end up being carriers. This is shown in the pedigree diagram below.

### X-linked recessive trait



**X-linked recessive pedigree diagram.** Source: dreamstime.com

In an X-linked recessive pedigree, much more males tend to be affected than females. With an affected male, the transmission of the condition tends to skip a generation from grandfather to grandson due to the inheritance pattern explained above. There is also no male-to-male transmission which is also explained above. For a female to be affected, she must have both an affected father and an affected or carrier mother, which is quite rare. If an affected female has offspring, all her sons will also be affected, as sons always inherit their X-chromosome from their mother and both of the mother's X-chromosomes would be affected in this situation. Examples of X-linked recessive conditions are haemophilia and Duchenne muscular dystrophy.

### Y-linked

In Y-linked inheritance, only males are affected as only males possess a Y chromosome. If the father has the condition, his sons will always have the condition too as the affected Y chromosome is inherited from the father:

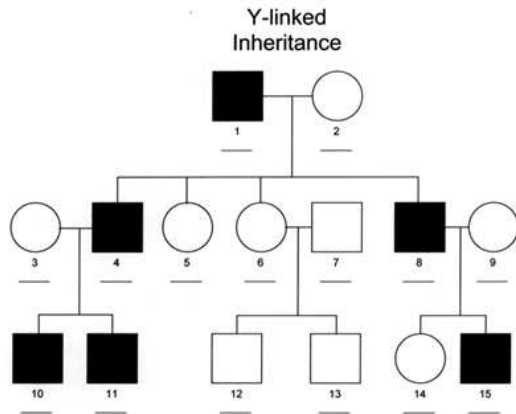
Parent genotypes:  $XY^d$   $XX$

Gametes:  $X$   $Y^d$   $X$   $X$

Possible genotypes:  $XY^d$   $XX$  (1:1)

|   | X  | $Y^d$  |
|---|----|--------|
| X | XX | $XY^d$ |
| X | XX | $XY^d$ |

**Y-linked recessive punnett square.** Source: SlimAcademy



**Y-linked pedigree diagram.** Source: *migr.org*

In a Y-linked pedigree, all sons with an affected father will have the condition. No females are affected as explained before. This is distinguishable from X-linked recessive inheritance because there is male-to-male transmission in Y-linked inheritance, but no male-to-male transmission in X-linked recessive.

Examples of Y-linked conditions are non-obstructive spermatogenic failure and webbed toes.

### Slim Summary!

- Pedigree diagrams are a useful way of visualising how different characteristics are inherited and passed on. Circles represent females, while squares represent males. A shaded circle or square means that the individual is affected;
- If a condition is X-linked dominant and the father is affected then all daughters will also be affected. You can often tell a condition is X-linked recessive as it is mostly found in males.

## Chapter 12 - Social and personal impact of living with a long-term health condition

### Introduction

This chapter discusses how chronic illness can affect far more than physical health, influencing a person's emotional wellbeing, social life, finances, and ability to work or study.

### Physical and Functional Limitations

Chronic fatigue, pain, or reduced mobility limit day-to-day activities such as walking, self-care, or driving. It can also lead to dependence on medications or assistive devices that may affect autonomy. Furthermore, frequent hospital visits and procedures can disrupt normal routines.

Clinical relevance: impacts ability to adhere to treatment and perform activities of daily living (ADLs); requires functional assessments and multidisciplinary support (physio, OT).

### Psychological and Emotional Effects

Depression and anxiety are common due to uncertainty, pain, or perceived loss of control. Altered self-image following amputation, visible disease, or weight changes. Fear of disease progression and death and coping fatigue from ongoing management and medical appointments may also have a big impact.

Clinical relevance: clinicians should screen for mood disorders and provide or refer for psychological support (CBT, counselling).

### Lifestyle Adjustments

Need for strict medication regimes, dietary restrictions, or lifestyle modifications can lead to disruption to hobbies, social activities, and sexual relationships. Furthermore, chronic fatigue may reduce participation in social and family life.

Clinical relevance: adherence challenges; importance of shared decision-making and realistic care planning.

### Financial Burden

The cost of long-term medication, equipment, and travel to appointments as well as possible job loss or reduced working capacity can be a big financial burden.

Clinical relevance: awareness of the role of social workers, occupational therapists, and financial support systems.

### Impact on Relationships

Role reversal within families (e.g., from caregiver to dependent) and emotional strain on partners and caregivers can lead to loss of intimacy or communication difficulties.

Clinical relevance: consider the impact on carers; involve family in care discussions; offer respite and support services.

### Social Isolation

Physical limitations or stigma may reduce social participation. Patients may avoid gatherings due to embarrassment (e.g., incontinence, visible illness). Chronic illness can cause withdrawal and loneliness.

Clinical relevance: assess social support and isolation risk; link to community or patient support groups.

### **Employment and Education**

Reduced ability to maintain employment due to fluctuating symptoms. Potential discrimination or lack of workplace accommodations or missed schooling or delayed career progression in younger patients.

Clinical relevance: encourage early occupational health involvement and discuss flexible work/study arrangements.

### **Stigma and Public Perception**

Some conditions, such as HIV, mental illness, or obesity, carry social stigma. Misunderstanding from others can cause shame, withdrawal, and reduced self-worth.

Clinical relevance: recognise stigma's effect on mental health and engagement with healthcare; foster empathy and non-judgmental communication.

### **Slim Summary!**

- Living with a long-term health condition affects many aspects of a person's life. They may struggle physically, emotionally, socially, and financially, their condition may also require many lifestyle adjustments - all of which affect the quality of life.

## Afterword

Phew, you did it! You've finished reading your summary. 🎉

Want to boost your confidence for the exam? Don't worry — we've got you covered! With one of our convenient subscriptions, you'll always receive your summaries at a discount and get them before anyone else. Also, we offer tons of practise questions and flashcards to ultimately prepare for you exams. Curious? Check out our website!


### Get in Touch with Slim Academy

Want to stay up to date with the latest developments at Slim Academy? Get in touch with us through:

 [www.slimacademy.nl](http://www.slimacademy.nl)

 klantenservice@slimacademy.nl

 @slimAcademy.nl

 010 214 32 45

We wish you lots of success with your studies and your exams!

### Team Slim Academy