Higher Human Biology
Unit 2 Physiology & Health Summary

1 Gamete production and fertilisation

(a) Gamete production in the testes.

- Sperm are produced in the testes in the seminiferous tubules.
- The interstitial cells of the testes produce the hormone testosterone.
- The prostate gland and seminal vesicles secrete fluids that maintain the mobility and viability of the sperm.

(b) Gamete production in the ovaries

- The ovaries contain immature ova in various stages of development.
- Each ovum is surrounded by a follicle that protects the developing ovum and secretes hormones.

(c) Fertilisation

Mature ova are released into the oviduct where they may be fertilised by sperm to form a zygote.

2 Hormonal control of reproduction

(a) Hormonal influence on puberty.

- The pituitary gland is stimulated to release follicle stimulating hormone (FSH), luteinising hormone (LH) or interstitial cell stimulating hormone (ICSH) by a releaser hormone produced in the hypothalamus.
- This triggers the onset of puberty

(b) Hormonal control of sperm production.

- FSH promotes sperm production and ICSH stimulates the production of testosterone.
- Testosterone also stimulates sperm production and activates the prostate gland and seminal vesicles.
- Negative feedback control of testosterone by FSH and ICSH.

(c) Hormonal control of the menstrual cycle

- The menstrual cycle takes approximately 28 days with the first day of menstruation regarded as day one of the cycle.
- FSH stimulates the development of a follicle and the production of oestrogen by the follicle in the follicular phase.
- Oestrogen stimulates proliferation of the endometrium preparing it for implantation, and affects the consistency of cervical mucus making it more easily penetrated by sperm. Peak levels of oestrogen stimulate a surge in the secretion of LH. This surge in LH triggers ovulation.
- Ovulation is the release of an egg (ovum) from a follicle in the ovary. It usually occurs around the mid-point of the menstrual cycle.

- In the luteal phase the follicle develops into a corpus luteum which secretes progesterone.
- Progesterone promotes further development and vascularisation of the endometrium preparing it for implantation if fertilisation occurs.

- The negative feedback effect of the ovarian hormones on the pituitary gland and the secretion of FSH and LH prevent further follicles from developing.
- The lack of LH leads to degeneration of the corpus luteum with a subsequent drop in progesterone levels leading to menstruation.
- If fertilisation does occur the corpus luteum does not degenerate and progesterone levels remain high.

3 The biology of controlling fertility
Infertility treatments and contraception are based on the biology of fertility.

(a) Women show cyclical fertility leading to a fertile period. Men show continuous fertility.

- Women are only fertile for a few days during each menstrual cycle.
- The fertile period can be identified by a woman's body temperature rising by around 0·5 °C after ovulation and her cervical mucus becoming thin and watery.
- Men continually produce sperm in their testes so show continuous fertility.

(b) Treatments for infertility

Stimulating ovulation
Ovulation is stimulated by drugs that prevent the negative feedback effect of oestrogen on FSH secretion. Other ovulatory drugs mimic the action of FSH and LH. These drugs can cause super ovulation that can result in multiple births or be used to collect ova for in vitro fertilisation (IVF) programmes.

Artificial insemination
Several samples of semen are collected over a period of time. Artificial insemination is particularly useful where the male has a low sperm count. If a partner is sterile a donor may be used to provide semen.

Intra-cytoplasmic sperm injection (ICSI)
If mature sperm are defective or very low in number, ICSI can be used. The head of the sperm is drawn into a needle and injected directly into the egg to achieve fertilisation.

In vitro fertilisation (IVF)
Surgical removal of eggs from ovaries takes place after hormone stimulation. Eggs are mixed with sperm in a culture dish. The zygotes, fertilised eggs are incubated until they have formed at least eight cells and are then transferred to the uterus for implantation.

IVF is used in conjunction with pre-implantation genetic diagnosis (PGD) to identify single gene disorders and chromosomal abnormalities.

(c) Physical and chemical methods of contraception.

Physical
Biological basis of physical methods used to prevent pregnancy is that barrier methods use a device to physically block the ability of sperm to reach the ova.

- Condom
- Diaphragm
- cervical cap
- Intra uterine device (IUD)
- Sterilisation procedures.

Chemical

- The oral contraceptive pill is a chemical method of contraception. It contains a combination of synthetic oestrogen and progesterone that mimics negative feedback preventing the release of FSH and LH from the pituitary gland.
- The progesterone-only (mini) pill causes thickening of the cervical mucus.
- The morning-after pill prevents ovulation or implantation.

4 Antenatal and postnatal screening
A variety of techniques can be used to monitor the health of the mother, developing fetus and baby.

(a) Antenatal screening
Antenatal screening identifies the risk of a disorder so that further tests and a prenatal diagnosis can be offered.

Ultrasound imaging
- Pregnant women are given two ultrasound scans.
- Dating scans which determine pregnancy stage and due date are used with tests for marker chemicals which vary normally during pregnancy. A dating scan takes place between 8 and 14 weeks.
- Anomaly scans between 18 and 20 weeks may detect serious physical abnormalities in the fetus.

Blood and urine tests
- Routine blood and urine tests are carried out throughout pregnancy to monitor the concentrations of marker chemicals.
- An atypical chemical concentration can lead to diagnostic testing to determine if the fetus has a medical condition.
- Measuring a chemical at the wrong time could lead to a false positive result.

Diagnostic testing
- Amniocentesis and chorionic villus sampling (CVS) are two diagnostic tests.
- Understand the advantages and disadvantages of their use. CVS can be carried out earlier in pregnancy than amniocentesis, although it has a higher risk of miscarriage.
- Cells from samples can be cultured to obtain sufficient cells to produce a karyotype to diagnose a range of conditions. A karyotype shows an individual's chromosomes arranged as homologous pairs.
- In deciding to proceed with these tests, the element of risk will be assessed, as will the decisions the individuals concerned are likely to make if a test is positive.

(b) Analysis of patterns of inheritance in genetic screening and counselling.
- Family histories can be drawn, analysed and interpreted over three generations to follow patterns of inheritance in genetic disorders.
- Patterns of inheritance include: autosomal recessive, autosomal dominant, incomplete dominance and sex-linked recessive single gene disorders.
- Standard genetic terms and their related symbols should be used — alleles, dominant, recessive, homozygous, heterozygous, carriers, genotype, phenotype, autosomes and sex chromosomes.

(c) Postnatal screening.
Diagnostic testing for phenylketonuria (PKU) takes place after birth. In PKU a substitution mutation means that the enzyme which converts phenylalanine to tyrosine is non-functional. Individuals with high levels of phenylalanine are placed on a restricted diet.

5 The structure and function of arteries, capillaries and veins

(a) Blood Circulation

Blood circulates from the heart through the arteries to the capillaries then to the veins and back to the heart. There is a decrease in blood pressure as blood moves away from the heart.

(b) The structure and function of arteries, capillaries and veins

- Structures include: endothelium, central lumen, connective tissue, elastic fibres, smooth muscle and valves.
- The endothelium lining the central lumen of blood vessels is surrounded by layers of tissue.
- Arteries carry blood away from the heart.
- Arteries have an outer layer of connective tissue containing elastic fibres and a middle layer containing smooth muscle with more elastic fibres.
- The elastic walls of the arteries stretch and recoil to accommodate the surge of blood after each contraction of the heart.
- To control blood flow, the smooth muscle surrounding arteries can contract causing vasoconstriction or relax causing vasodilation. This process allows changing demands of the body's tissues to be met.
- Capillaries allow exchange of substances with tissues through their thin walls.
- They are only one cell thick so allow quick and efficient exchange of materials.
- Veins carry blood towards the heart
- Veins have an outer layer of connective tissue containing elastic fibres but a much thinner muscular wall than arteries. Blood flows along veins at lower pressure than arteries
- The lumen of a vein is wider than that of an artery
- Blood flows along veins at lower pressure than arteries so veins contain valves to prevent the backflow of blood.

(c) The exchange of materials between tissue fluid and cells through pressure filtration and the role of lymphatic vessels.

- Blood is carried to the tissues in thick walled arteries which, once they enter an organ they divide into many arterioles which, again divide into capillaries
- Pressure filtration causes plasma to pass through capillary walls into the tissue fluid surrounding the cells. Fluid contains small soluble molecules but not the blood cells and large plasma protein molecules
- Tissue fluid supplies cells with glucose, oxygen and other substances which diffuse down concentration gradients
- Carbon dioxide and other metabolic wastes diffuse out of the cells and into the tissue fluid to be excreted.
- At the venule end of the capillary bed tissue fluid enters the capillaries again by osmosis as the blood cells and plasma proteins cause a water concentration gradient
- Much of the tissue fluid returns to the blood.
- Lymphatic vessels absorb excess tissue fluid and return it as lymph to the circulatory system.
- Lymph is returned to the blood then the heart by skeletal muscles squeezing it past the next valve
- Tissue fluid and blood plasma are similar in composition with the exception of plasma proteins, which are too large to be filtered through the capillary walls.

6 The structure and function of the heart

Blood flow through the heart and its associated blood vessels.

(a) Cardiac output and its calculation.

The volume of blood pumped through each ventricle per minute is the cardiac output.
Cardiac output is determined by heart rate and stroke volume

- The stroke volume is the volume of blood pumped out of the heart with each heartbeat.
- The pulse rate corresponds to the heart rate

An individual's cardiac output can be worked out by multiplying the heart rate by the stroke volume

(CO = HR x SV).

The left and right ventricles pump the same volume of blood through the aorta and pulmonary artery.

(b) The cardiac cycle.

Functions of diastole, atrial systole and ventricular systole.

- The right side collects deoxygenated blood from the body and pumps it to the lungs to collect oxygen
- The left side collects oxygenated blood from the lungs and pumps it to the body
- The walls of the heart are made of cardiac muscle which can contract rapidly, without fatigue for a life time
- During diastole, blood returning to the atria flows into the ventricles
- The build-up of pressure forces open the atrio-ventricular valves and blood flows into the ventricles.
- Atrial systole transfers the remainder of the blood through the atrio-ventricular (AV) valves to the ventricles.
- Once full the ventricles muscular walls contract and this Ventricular systole closes the AV valves and pumps the blood out through the semi lunar (SL) valves to the aorta and pulmonary artery and then to the lungs
- In diastole, the higher pressure in the arteries closes the SL valves.

Effect of pressure changes on atrio-ventricular (AV) and semi lunar (SL) valves

- Pressure changes cause the opening and closing of the AV and SL valves which are responsible for the heart sounds heard with a stethoscope.

(c) The structure and function of the cardiac conducting system

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- The heartbeat originates in the heart itself.
- The sino-atrial node (SAN) controls the timing of cell contraction
- The auto-rhythmic cells of the sino-atrial node (SAN) or pacemaker, located in the wall of the right atrium, set the rate at which the heart contracts.
- The timing of cardiac muscle cell contraction is controlled by impulses from the SAN spreading through the atria causing atrial systole to the atrio-ventricular node (AVN), located in the centre of the heart
- The atrio-ventricular node (AVN) is important for signal transmission.
- Impulses from the AVN travel down fibres in the central wall of the heart and then up through the walls of the ventricles, causing ventricular systole.

Impulses in the heart generate currents that can be detected by an electrocardiogram (ECG). ECGs can be used to calculate heart rate and link the waves to atrial systole, ventricular systole and diastole.

The medulla regulates the rate of the sino-atrial node through the antagonistic action of the autonomic nervous system (ANS).

- A sympathetic nerve releases noradrenaline which increases the heart rate
- A parasympathetic nerve releases acetylcholine which decreases the heart rate.

(d) Blood pressure changes in the aorta during the cardiac cycle.

- Blood pressure increases during ventricular systole and decreases during diastole.
- A sphygmomanometer is used to measure blood pressure.
- An inflatable cuff stops blood flow, in the artery, and deflates gradually. The blood starts to flow (detected by a pulse) at systolic pressure. The blood flows freely through the artery (and a pulse is not detected) at diastolic pressure.
- A typical blood pressure reading for a young adult is 120/80 mmHg.
- Hypertension (high blood pressure) is a major risk factor for many diseases including coronary heart disease.

7 Pathology of cardiovascular disease (CVD)

(a) Process of atherosclerosis, its effect on arteries and blood pressure.

- Atherosclerosis is the accumulation of fatty material (consisting mainly of cholesterol, fibrous material and calcium) forming an atheroma or plaque beneath the endothelium.
- As the atheroma grows the artery thickens and loses its elasticity.
- The diameter of the lumen becomes reduced and blood flow becomes restricted resulting in increased blood pressure.
- Atherosclerosis is the root cause of various cardiovascular diseases (CVD) — angina, heart attack, stroke and peripheral vascular disease.

(b) Thrombosis

- Atheromas may rupture damaging the endothelium.
- The damage releases clotting factors that activate a cascade of reactions resulting in the conversion of the enzyme prothrombin to its active form thrombin.
- Thrombin causes molecules of the plasma protein fibrinogen to form threads of fibrin.
- The fibrin threads form a meshwork that clots the blood, seals the wound and provides a scaffold for the formation of scar tissue.
- The formation of a clot (thrombus) is referred to as thrombosis.
- In some cases a thrombus may break loose forming an embolus which travels through the bloodstream until it blocks a blood vessel.
- A thrombosis in a coronary artery may lead to a myocardial infarction (MI), commonly known as a heart attack.
- A thrombosis in an artery in the brain may lead to a stroke.
- Cells are deprived of oxygen leading to death of the tissues.

(c) Causes and effects of peripheral vascular disorders.

Peripheral vascular disease is narrowing of the arteries due to atherosclerosis of arteries other than those of the heart or brain. The arteries to the legs are most commonly affected. Pain is experienced in the leg muscles due to a limited supply of oxygen.

A deep vein thrombosis (DVT) is a blood clot that forms in a deep vein, most commonly in the leg. This can break off and result in a pulmonary embolism in the lungs.

(d) Control of cholesterol levels in the body.

- Cholesterol is a type of lipid found in the cell membrane.
- Cholesterol is also used to make the sex hormones — testosterone, oestrogen and progesterone.
- Cholesterol is synthesised by all cells, although 25% of total production takes place in the liver.
- A diet high in saturated fats or cholesterol causes an increase in cholesterol levels in the blood.
- There are two types of lipoproteins with different roles -high density lipoproteins (HDL) and low density lipoproteins (LDL).
- HDL transports excess cholesterol from the body cells to the liver for elimination. This prevents accumulation of cholesterol in the blood.
- LDL transports cholesterol to body cells.
- Most cells have LDL receptors that take LDL into the cell where it releases cholesterol.
- Once a cell has sufficient cholesterol a negative feedback system inhibits the synthesis of new LDL receptors and LDL circulates in the blood where it may deposit cholesterol in the arteries forming atheromas.
- Ratios of HDL to LDL are important in maintaining health.
- A higher ratio of HDL to LDL will result in lower blood cholesterol and a reduced chance of atherosclerosis.
- Regular physical activity is of benefit because it tends to raise HDL levels.
- Dietary changes aim to reduce the levels of total fat in the diet and to replace saturated with unsaturated fats are also of benefit.
- Prescribed medications, drugs such as statins reduce blood cholesterol by inhibiting the synthesis of cholesterol by liver cells.

8 Blood glucose levels and obesity

(a) Chronic elevated blood glucose levels lead to atherosclerosis and blood vessel damage.

- Chronic elevation of blood glucose levels (e.g. due to untreated diabetes) leads to the endothelium cells taking in more glucose than normal, damaging the blood vessels.
- Atherosclerosis may develop leading to cardiovascular disease, stroke or peripheral vascular disease (blood vessels leading to hands, arms, toes, feet, legs).
- Small blood vessels damaged by elevated glucose levels may result in haemorrhage of blood vessels in the retina, renal failure or peripheral nerve dysfunction (nerves leading to hands, arms, toes, feet, legs).

(b) Pancreatic receptors and the role of hormones in negative feedback control of blood glucose through insulin, glucagon and adrenaline.

- Glucose is the substrate for respiration, and so the blood glucose concentration is maintained within fine limits
- Blood glucose concentration is monitored in the pancreas
- The pancreas controls blood glucose with two hormones, insulin and glucagon, which act antagonistically
- Pancreatic receptors respond to raised blood glucose levels by increasing secretion of insulin from the pancreas. The high concentration of insulin makes the liver cells more permeable to glucose so they absorb more
- Insulin activates the conversion of glucose to glycogen in the liver decreasing blood glucose concentration.
- Pancreatic receptors respond to lowered blood glucose levels by increasing secretion of glucagon from the pancreas.
- Glucagon activates the conversion of glycogen to glucose in the liver increasing blood glucose concentration.
- During exercise and fight or flight responses, glucose concentrations in the blood are raised by adrenaline, released from the adrenal glands, stimulating glucagon secretion and inhibiting insulin secretion.
- Once the emergency is over insulin brings the blood glucose concentration back down to normal
- The regulation of blood glucose level is an example of negative feedback control

(c) Type 1 and type 2 diabetes

- Type 1 diabetes usually occurs in childhood.
- A person with type 1 diabetes is unable to produce insulin and can be treated with regular doses of insulin.

- Type 2 diabetes typically develops later in life. The likelihood of developing type 2 diabetes is increased by being overweight.
- In type 2 diabetes, individuals produce insulin but their cells are less sensitive to it. This insulin resistance is linked to a decrease in the number of insulin receptors in the liver, leading to a failure to convert glucose to glycogen.

- In both types of diabetes, individual blood glucose concentrations will rise rapidly after a meal.
- The kidneys will remove some of this glucose resulting in glucose appearing in urine. Testing urine for glucose is often used as an indicator of diabetes.

Diagnosis
The glucose tolerance test is used to diagnose diabetes. The blood glucose concentrations of the individual are initially measured after fasting. The individual then drinks a glucose solution and changes in their blood glucose concentration are measured for at least the next two hours.

The blood glucose concentration of a diabetic usually starts at a higher level than that of a non-diabetic.

During the test a diabetic's blood glucose concentration:
- increases to a much higher level than that of a non-diabetic
- takes longer to return to its starting concentration.

(d) Obesity
- Obesity is a major risk factor for cardiovascular disease and type 2 diabetes.
- Obesity is characterised by excess body fat in relation to lean body tissue such as muscle.
- Obesity may impair health.

Body mass index (BMI)
- BMI is commonly used to measure obesity but can wrongly classify muscular individuals as obese.
- BMI = body mass divided by height squared (BMI (kg m^{-2}) = weight (kg) / height $(m)^2$.).
- A BMI greater than 30 is used to indicate obesity.

Role of diet and exercise in reducing obesity and cardiovascular disease (CVD).
- Obesity is linked to high fat diets and a decrease in physical activity.
- The energy intake in the diet should limit fats and free sugars, as fats have a high calorific value per gram and free sugars require no metabolic energy to be expended in their digestion.
- Exercise increases energy expenditure and preserves lean tissue.

- Exercise can help to reduce risk factors for CVD:
    - by keeping weight under control,
    - minimising stress,
    - reducing hypertension
    - improving blood lipid profiles.