Circulatory systems

Closed circulatory systems:

- Blood leaves heart under pressure # arteries # arterioles # capillaries
- Capillaries come in large numbers. They exchange substances between the blood and cells.
- After passing through capillaries # blood goes back to the heart via venules # veins
- Valves in the veins ensure that blood only flows in one direction.

Single circulatory system: (e.g. in fish)

- Heart pumps deoxygenated blood # gills
- Gaseous exchange (diffusion of CO_2 from blood to H_2O that surrounds the gills, and diffusion of O_2 from H_2O into the blood)
- Blood leaves gills # rest of body # heart

Double circulatory system:

- Right ventricle pumps deoxygenated blood to the lungs where it receives oxygen.
- The oxygenated blood then returns to the heart to be pumped a second time (by the left ventricle) out to the rest of the body.

Circulation

In the circulatory system all the particles it contains are transported in one direction in a process known as mass flow.

properties of water that make it a good transport medium:

- Water is a very good transport medium.
- Water is liquid at room temperature.
- The hydrogen's in the water push away from each other making the molecule V shaped.
- many water molecules can bond together forming hydrogen bonds, as the negatively charged oxygen of one molecule bonds to the positively charged hydrogen of another.
- The hydrogen bonding holds them together and results in many of the properties of water.
- Many chemicals dissolve easily in water.
- Polar molecules (hydrophilic) dissolve easily in water.
- Non-polar (hydrophobic) substances, e.g. lipids, do not dissolve in water.
- Water has a high boiling point because the hydrogen bonds require a lot of energy to break as they are very strong.

The heart and blood vessels

The heart consists of:

- Aorta (to body)
- Pulmonary artery (to lungs)
- Pulmonary veins (from lungs)
- Left and right atrium
- Left and right ventricle
- Atrioventricular valves (separates the ventricles and atrium)
- Semi-lunar valve (separates the ventricles from the aorta)
- Inferior vena cava (from lower body)
- Superior vena cava (from head and arms)

Arteries and veins:

- Arteries: narrow lumen Veins: wide lumen
- Arteries: thick walls Veins: thinner walls
- Arteries: more collagen, elastic fibers and smooth muscle Veins: has less
- Arteries: no valves Veins: have valves

Capillaries: Are only one cell think and join the small arteries (arterioles) and small veins (venules)

How does blood move through the vessels?

- Every time the heart contracts (systole) blood is forced into arteries and their walls stretch to accommodate the blood.
- During relaxation of the heart (diastole) the elasticity of the walls causes them to recoil behind the blood pushing the blood forward.

How the valves in veins work:

- Blood passed through the vein
- skeletal muscles contract
- contraction pushes blood forward toward the heart through the open valves
- valve behind is closed preventing the back-flow of blood
- when the blood has passed through the open valve the muscles relax
- this causes the vales to shut behind it again preventing back-flow

The heart needs a constant supply of blood. Two vessels called the coronary arteries give the heart with a constant supply of blood.

How the heart works

The four chambers of the heart are continually and relaxing in a sequence known as the cardiac cycle. Contraction of a chamber is systole and relaxation diastole.

Phase 1: Atrial systole

- blood under low pressure flows into the left and right atria from the pulmonary veins and vena cava.
- as atria fill pressure against atrioventricular valves pushes them open and blood starts leaking into the ventricles.
- the atria walls then contract forcing more blood into the ventricles.

Phase 2: Ventricular systole

- Ventricles contract from base upwards increasing the pressure.
- this pushes blood up and out through the arteries.
- the pressure of blood against the atrioventricular valves closes them and prevents and prevents back-flow into the atria.

How the heart works continued

Phase 3: Diastole

- Atria and ventricles relax during diastole.
- Elastic recoil lowers pressure in the atria and ventricles.
- Blood under higher pressure in the arteries is drawn back towards the ventricles, closing the semi-lunar valves.
- The coronary arteries fill during diastole.
- Low pressure in the atria helps draw blood into the heart from the veins.

Closing the atrioventricular valves and then the semi-lunar valves creates the characteristic sounds of the heart.

What is atherosclerosis?

Atherosclerosis can lead to coronary heart disease and strokes. It does this by blocking an artery with fatty deposits .

Stages of atherosclerosis:

- Endothelium becomes damaged (e.g. due to high blood pressure, or cigarette smoke)
- Damage causes inflammatory response. White blood cells move into the artery wall, they accumulate chemicals from the blood (cholesterol)
- A deposit then builds up called an atheroma
- Calcium salts and fibrous tissue build up at site and form a hard swelling (plaque)
- This makes the artery lose some of its elasticity (hardens)
- It also causes the artery to narrow
- This makes it difficult for the heart to pump blood around the body and results in high blood pressure
- Positive feedback builds up as the increased blood pressure makes it more likely that more plaques will form.

Blood clot

When blood vessel walls are damaged a blood clot is likely to form.

- Platelets come into contact with vessel wall and change from flat discs to spheres with long thin projections.
- this change causes them to stick to the exposed collagen in the wall and each other and from a temporary platelet plug.
- they also release substances that activate more platelets
- the contact of blood with collagen causes chemical changes in the blood:
  - Soluble plasma protein **prothrombin**
  - Converted into **thrombin** an enzyme
  - thrombin catalyses the conversion of another soluble plasma protein called **Fibrinogen**
  - into long insoluble strands of the protein **fibrin**
  - The fibrin strands from a mesh that traps blood cells to form the clot.

Aneurysms can also form which are a build up of blood behind a narrowed part of an artery.

Identifying risk factors for CVD

Large-scale studies have been undertaken to find the risk factors for many common diseases including cardiovascular disease.

There are two common studies:

Cohort study - a group of people are followed over time to see who develops the disease

Case-control studies - a group of people who have the disease are compared with a group who do not have the disease

Features of a good study:

- clear aim
- representative sample
- valid and reliable results

Identifying risk factors for CVD continued

Rick factors that increase the chance of getting CVD:

- High blood pressure
- Obesity
- Blood cholesterol and other dietary factors
- Smoking
- Genetic inheritance

The risk of CVD is higher for men than women in the UK.

The risk of getting CVD also increases with age.

High blood pressure: (hypertension)

blood pressure is measured by using a sphygmomanometer, it measures the systolic pressure and the diastolic pressure in the arteries.

it is shown by placing systolic pressure over diastolic pressure. In a healthy person a systolic pressure of between 100 and 140 is expected and a diastolic pressure between 60 and 90.

Sugars

Mono-saccharides are single sugar units.

They can be joined in condensation reactions to form disaccharides which are made of two mono-saccharides.

They can also be joined together to form polysaccharides which contain three or more sugar units.

When two sugars join together via carbon 1 and carbon 4 on another a glycosidic bond is formed and produces water.

Sugars: (disaccharides)

- Sucrose - (**glucose + fructose**) form in which sugar is transported in a plant
- Maltose - (**glucose + glucose**) produced when amylase breaks down starch
- Lactose - (**galactose + glucose**) the sugar found in milk

The glycosidic bond between the two sugars can be broken by hydrolysis, which is the addition of water.

There are three main polysaccharides that are found in food: starch and cellulose in plants, and glycogen in animals. Starch and glycogen act as energy storage molecules within cells. They are suitable for storage because they are compact molecules.

Starch, glycogen and cellulose

Starch is made up of a mixture of two molecules, **amylose** and **amylopectin**. Starch is a major source of energy and is found in many foods such as fruit and vegetables.

Amylose:

- straight chain between 200 and 500 glucose molecules in length
- 1,4 glycosidic links between adjacent glucose molecules
- chain is coiled into a spiral shape

Amylopectin:

- polymer of glucose with side branches
- 1,6 glycosidic links
- has side branches

Cellulose is known as dietary fibre. It has an important function as it helps the movement of material through the digestive tract.

Glycogen is stored by bacteria, fungi and animals. Its side branches mean that it can be rapidly hydrolysed giving easy access to stored energy. (Humans store in liver and muscles)

Lipids

Lipids enhance the flavour and palatability of food.

They are insoluble in water but soluble in organic solvents such as ethanol.

The most common lipids we eat are triglycerides:

- used as energy stores in plants and animals
- they are made up of three fatty acids and one glycerol molecule
- these are linked together by condensation reactions
- the bond that forms between them is called an ester bond
- three ester bonds are formed in a triglyceride

Saturated fats: if the fatty acid chains in a lipid contain the maximum number of hydrogen atoms they are saturated. There are also no double bonds.

Unsaturated fats: Monounsaturated fats have one double bond between two of the carbon atoms in the chain. Polyunsaturated have a larger number of double bonds.

Another lipid is cholesterol which is essential for good health, although too much of it can lead to a high blood cholesterol level and can be bad for us.

The energy balance

A constant supply of energy is needed to maintain your essential body processes (e.g. pumping the heart). The amount of energy needed for this is called the **basal metabolic rate** (BMR)

BMR is higher in:

- Males
- Heavier people
- Younger people
- More active people

Body mass index (BMI) is a way of classifying body weight relative to a persons height. A BMI of under 20 is considered to be underweight, a BMI of over 40 is considered to be severely obese.

Calculating BMI:

BMI = weight in kg / height^2 in m

Cholesterol

Cholesterol is not soluble in water. To be transported in the bloodstream it is combined with proteins to form soluble **lipoproteins**.

There are two main types of **lipoprotein** these are:

Low density lipoprotein (LDL's):

- main cholesterol carrier in the blood
- triglycerides from saturated fats in our diet combine with the cholesterol and protein to form LDL's
- they circulate in the blood stream and bind to receptor sites on cell membranes
- they are consist of more cholesterol than protein

High density lipoproteins (HDL's):

- are made of more protein than cholesterol hence the high density
- are made when tryglycerides fro unsaturated fats combine with cholesterol and protien
- they transport cholesterol from the body tissues to the liver where it is broken down
- it helps lower blood cholesterol and helps remove plagues in arteries

Smoking and inactivity

Smoke affects the circulatory system in the following ways:

- The haemoglobin in the red blood cells carries carbon monoxide instead of oxygen, reducing the amount of oxygen that gets to the cells
- Nicotine in smoke stimulates the production of the hormone adrenaline. This hormone causes an increase in heart rate and the arteries and arterioles to constrict.
- the chemicals that are found in smoke can cause damage to yhe linine of the arteries
- smoking has also been linked with a reduction in HDL cholesterol levels

Inactivity can also increase the chances of getting CVD:

- as increased exercise can help prevent high blood pressure and can also help lower it
- it raises HDL cholesterol level without affecting the LDL levels
- can help you survive a stroke or heart attack

Apolipoprotein

Apolipoproteins are the protein component of lipoproteins (found in liver and intestines) there are several types of Apolipoproteins:

Apolipoprotein A (APOA):

- main protein in HDL that helps remove cholesterol into the liver for excretion
- mutations in apoA gene result in low HDL levels and reduced removal of cholesterol from the blood

Apolipoprotein B (APOB):

- main protein in LDL that transfers cholesterol from blood to cells
- mutations in apoB gene cause higher levels of LDL in the blood

Apolipoprotein E (APOE):

- main component of HDL's and very low density lipoproteins (VLDL's)
- involved in removal of excess cholesterol from lipoproteins from the blood to the liver
- aopE has three common alleles that produce three forms of the protein, E2, E3, and E4
- APEO4 slows removal of cholesterol from the blood

Other risk factors for CVD

The role of antioxidants:

high levels of antioxidants protect against heart disease.

Salt:

too much salt can cause the kidneys to retain water. higher fluid levels in the blood result in elevated blood pressure.

Stress:

in stressful situations the release of adrenaline causes arteries and arterioles to constrict resulting in raised blood pressure

Alcohol:

heavy drinking raises blood pressure, contributes to obesity, and can cause an irregular heartbeat.

Reducing the risk of CVD

The risk of getting CVD can be reduced by:

- stopping smoking
- maintaining a blood pressure below 140/85 mmHg
- maintaining a low blood cholesterol level
- maintaining a normal BMI/ low waist-to-hip ratio
- taking more exercise
- moderate use of alcohol

Ways of controlling blood pressure:

- ACE inhibitors
- calcium channel blockers
- Diuretics

Reducing blood cholesterol levels:

Mainly the drug statins are used. They inhibit an enzyme involved in the production of LDL cholesterol.

Changing diet to reduce the risk of CVD

This would include:

- Energy balanced
- Reduced saturated fat
- More polyunsaturated fats
- Reduced cholesterol
- Reduced salt
- More non-starch polysaccharides (pectins and guar gum)
- Oily fish (contain omega-3 fatty acids)
- More fruit and vegetables (contain antioxidants)
- Include functional foods containing sterols and stanols

Anticoagulant and platelet inhibitory drug treatment:

The tenancy for platelet aggregation and clotting is reduced by platelet inhibitory drugs and anticoagulant drugs.

Cystic fibrosis (CF)

CF problems:

- CF is caused by a faulty transport protein in the surface membranes of epithelial cells
- CF creates a sticky mucus layer that lines the tubes and ducts in the gas exchange, digestive and reproductive systems.
- The sticky mucus increases the chances of lung infection and makes gas exchange less efficient.
- microorganisms become trapped in the sticky mucus causing illness
- cilia cannot move the mucus because it is too sticky
- low levels of oxygen in the mucus so harmful bacteria can live in these conditions
- gasses such as oxygen cross the walls of the alveoli into the blood system by diffusion, the sticky mucus makes it harder for diffusion to take place

Gas exchange

Features of a good gas exchange surface:

- large surface area for the alveoli
- numerous capillaries around the alveoli
- thin walls of the alvioli and capillaries meaning a short distance between the alveolar air and the blood in the capillaries

The rate of diffusion depends on:

- **surface area** - rate of diffusion is directly proportional to the surface area
- **concentration gradient** - rate of diffusion is directly proportional to the difference in concentration across the gas exchange surface.
- **thickness of the gas exchange surface** - the rate of diffusion is inversely proportional to the thickness of the gas exchange surface. (the thicker the surface the slower the rate of diffusion)

Why is CF mucus so sticky?

The mucus on the surface of the epithelial cells is sticky because it contains less water than normal.

Protein structure is key in protein function:

Amino acids consist of an amine group, a carboxylic acid group,

Primary structure - Two amino acids join in a condensation reaction to form a dipeptide (this can be more than two amino acids in length and they are joined by a peptide bond)

Secondary structure - The chain of amino acids twist to form an alpha-helix, hydrogen bonds form between the C=O of the carboxylic acid and the -NH of the amine group of different amino acids. Several chains my link together forming a beta-pleated sheet.

Tertiary and quaternary structure - a polypeptide chain bends and folds to produce a three dimensional shape (maintained by interactions between R groups)

A protein may be made up of several polypeptide chains held together this is known as the quaternary structure.

Proteins

Proteins are divided into two groups:

- globular proteins
- fibrous proteins

Globular proteins:

- folded into a compact spherical shape
- soluble because of hydrophilic side chains that are on the outside of them
- enzymes are globular proteins
- their three-dimensional shape is critical to their roles in binding to other substances
- e.g. of a globular protein is transport proteins within membranes

Fibrous proteins:

- are long chains
- several polypeptide chains can be cross linked for additional strength
- they are insoluble
- e.g. of a fibrous protein is keratin found in the hair and skin

Cell membrane structure

Phospholipid bilayer:

- The cell surface membrane consists of a bilayer that contains two layers of phospholipids.
- In a phospholipid there are two fatty acids and a negatively charged phosphate group.
- the phosphate head of he molecule is polar one end is slightly positive and the other negative
- so this makes the phosphate head attract other polar molecules like water so it is hydrophilic
- fatty acid tails are non-polar so they are hydrophobic

Cells are filled with watery cytoplasm and are surrounded by aqueous tissue fluid. so the phospholipids form a bilayer. This stops the hydrophobic fatty acids tails from being in contact with the water on both sides of the membrane and ensures that the hydrophilic phosphate heads are in contact with the water.

Fluid mosaic model

The cell surface membrane consists of:

- Phospholipid bilayer (all the molecules are arranged so that the hydrophobic tails are inside the bilayer)
- proteins
- cholesterol
- glycoproteins (protein molecules with polysaccharides attached)
- glycolipids (lipid molecules with polysaccharides attached)
- Channel proteins

Why is it called the fluid mosaic model?:

The plasma membrane is described to be fluid because of the lipids and membrane proteins that can move freely.

The membrane is called mosaic because like a mosaic that is made up of many different parts so is the plasma membrane.

How do substances pass through cell membranes?

Molecules and ions move across membranes by:

- diffusion
- osmosis
- active transport
- exocytosis
- endocytosis

Diffusion: (or passive transport)

Diffusion is the net movement of molecules from a region where they are at a higher concentration to a region of their lower concentration. (diffusion continues until equilibrium)

Faciliated diffusion:

Is when molecules and ions that are larger than carbon dioxide cross the membrane with the aid of proteins. They diffuse through water-filled pores within channel proteins.

Some proteins that help in this are called carrier proteins, the ion or molecule binds onto a specific site on the protein, the protein changes shape and then the ion or molecvule can cross the membrane.

Transport across the cell membrane

Diffusion :

- High to low concentration until equilibrium reached
- hydrophobic (lipid - soluble) or small uncharged molecules
- through phospholipid bilayer
- passive, no energy required

Facilitated diffusion:

- high to low concentration until equilibrium reached
- hydrophilioc molecules or ions
- through channel proteins or via carrier proteins that change shape
- passive, no energy required

Osmosis:

- a type of diffusion involving movement of free water molecules
- high to low concentration of free water molecules untill equilibrium reached
- through phospholipid bilayer
- passive, no energy required

Transport across the cell membrane

Active transport:

- against a concentration gradient, low to high concentration
- through carrier proteins that change shape
- requires energy

Exocytosis:

- used for bulk transport of substances out of the cell
- vesicles fuse with the cell surface membrane, releasing their contents

Endocytosis:

- used for bulk transport of substances into the cell
- vesicles are created from the cell surface membrane, bringing their contents into the cell

Regulating water in the mucus in affected lungs

With excess water:

- Na+ is actively pumped across the basal membrane
- Na+ diffuses through sodium channels in the apical membrane
- Cl- diffuses down electrical gradient
- water is drawn out of cells by osmosis due to the high salt concentration in the tissue fluid
- water is drawn out of the mucus by osmosis

With too little water:

- Cl- s pumped into the cell across the basal membrane
- Cl- diffuses through the open CFTR channels
- Na+ diffuses down the electrical gradient into the mucus
- elevated salt concentration in the mucus draws water out of the cell by osmosis
- water is drawn out of the cell by osmosis

With CF: CTFR channel is absent or not functioning, and the Na+ channel is permanently open, water is continually removed from the mucus by osmosis.

Effect of CF on digestive system

- In a person with CF the pancreatic duct becomes blocked by sticky mucus, impairing the release of digestive enzymes.
- The lower concentration of enzymes within the small intestine reduces the rate of digestion.
- Because of this the food is not fully digested and not all the nutrients can be absorbed
- The pancreatic enzymes can also become trapped behind the mucus blocking the pancreatic duct.
- The enzymes damage the pancreas.
- Damage to the cell walls in the pancreas that produce insulin, insulin is involved in the control of blood sugar levels, a form of diabetes can be the result.

Enzymes

Enzymes are globular proteins that act ad biological catalysts (They speed up chemical reactions). There is a depression on the surface of the enzyme molecule called the active site.

Lock and Key theory:

- a molecule with a complimentary shape can fit into the active site
- the substrate molecules form temporary bonds with the amino acids of the active site to produce an enzyme-substrate complex
- the enzyme holds the substrate molecules in a way that they react more easily
- when the reaction is finished the products are released, leaving the enzyme unchanged
- the substrate is known as the 'key' which fits into the enzymes 'lock'

Induced fit theory:

- active site is flexible
- when the substrate enters the active site the enzyme molecule changes shape slightly fitting more closely round the substrate

Effect of CF on reproductive system

Females with CF:

- have a reduced chance of becoming pregnant because a mucus plug develops in the cervix this stops sperm from reaching the egg

Males with CF:

- commonly lack the vas deferens (sperm duct) on both sides which means sperm cannot leave the testes.
- When the vas deferens is present it can become blocked by a thick sticky mucus layer, which means fewer sperm are present in each ejaculate

The structure of DNA

Gene and genome:

a gene is a sequence of bases on a DNA molecule coding for a sequence of amino acids in a polypeptide chain. Together all genes in a individual are known as the genome.

DNA is a chain of nucleotides:

DNA is one type of nucleic acid, called deoxyribonucleic acid. It is a long chain molecule made up of many units called nucleotides or mononucleotides

a mononucleotide consists of:

- a deoxyribose sugar
- a phosphate
- a base

A mononucleotide contains three molecules linked together by condensation reactions. They link together by condensation reactions between the sugar of one and the phosphate of the next.

Protein synthesis

Transcription: (in the nucleus)

- DNA double helix unwinds and hydrogen bonds break
- the template strand is used in the production of a messenger RNA molecule
- every triplet code on DNA gives rise to a complimentary codon on messenger RNA
- the completed messenger RNA molecules now leaves the nucleus through a pore in the nuclear envelope
- and enters the cytoplasm

Translation: (on ribosomes)

- ribosomes are small organelles made of ribosomal RNA and protein
- ribosomes are found free in the cytoplasm or attached to endoplasmic reticulum
- a transfer RNA molecule carrying an amino acid has three bases called an anticodon
- these pair with complimentary bases on the mRNA codon
- then the amino acids that the tRNA carry are joined by peptide bonds

DNA

There are four bases:

- adenine (A)
- cytosine (C)
- guanine (G)
- thymine (T)

In DNA there are two long strands of nucleotides twisted around to form a double helix. The sugars and phosphates are on the outside, and the bases point inwards and are held together by hydrogen bonds.

The bases pair up along the two nucleotides A pairs up with G and T with C.

The code carried by the DNA is a three-base or triplet code, each group of three bases codes for an amino acid there are also start and stop signals.

DNA and RNA:

- RNA has a single strand made of a string of RNA nucleotides.
- RNA contains a ribose sugar instead of a deoxyribose sugar
- In RNA U (uracil) replaces T (the RNA never contains thymine

DNA replication

When a cell divides an exact copy of the DNA must be produced so that each of the daughter cells receives a copy This process of copying the DNA is called replication.

There are three possible ways DNA could replicate:

- fragmentary replication - all DNA strands are made up of a mixture of original parent DNA nucleotides and new nucleotides
- conservative replication - one DNA molecule has two original parent DNA strands, the other molecule has two new strands
- semi conservative replication - each DNA molecule contains one original parent strand and one new strand

If a test tube containing DNA dissolved in a special density gradient solution is centrifuged. Heavy DNA containing N-15 sinks to the bottom, light DNA containing N-14 collects in a band near the top and DNA of a medium density (containing heave and light nucleotides) is in the middle.

What goes wrong with DNA?

Mistakes in replication:

Sometimes DNA replication does not work properly. As the new strand of DNA is being built and incorrect base may slip into place.

Sickle cell anemia:

mutation in the gene that codes for one of the polypeptide chains in haemoglobin (the pigment in red blood cells which carries oxygen around the body.

The base adenine replaces thymine at one position along the chain. A protein valine is made instead of glutamic acid. The red blood cells are distorted and the sickle shaped cells carry less oxygen and can block blood vessels.

Cystic fibrosis:

Mutations affect CFTR protein in different ways.

How genes are passed on

Genes and chromosome pairs:

Every cell contains two copies of each gene, one from each parent.We have 23 pairs of chromosomes and the chromosomes in each pair are called homologous chromosomes. In each pair one comes from the mother and one from the father.

Genotypes, phenotypes and alleles:

There is a normal allele which codes for the functioning CFTR protein (F). And there is a mutated allele which produces a non-functional protein (f).

FF - a person with two identical copies of the normal allele does not have cystic fibrosis

ff - a person with two copies of the mutated allele has cystic fibrosis

Ff - a person with one normal allele and one mutated allele does not have cystic fibrosis but is a carrier and could have children with the disease.

The alleles that a person has make up their genotype.

Treatments for CF

Medication:

- Bronchodilators - inhaled using a nebuliser. the drugs relax the muscles in the airways opening them up.
- Antibiotics - used to kill or prevent growth of bacteria in the lungs
- DNAase enzymes - DNAase enzymes can be inhaled using a nebuliser. They break down the DNA, so the mucus is easier to clear from the lungs.
- Steroids - used to reduce inflammation of the lungs.

Diet: eat high energy foods, and double the quantity of protein compared to people without CF, and salt supplements.

Digestive enzyme supplements: Taking these with food helps to complete the process of digestion.

Physiotherapy: Rhythmical tapping of the walls of the chest cavity can help loosen the mucus and improve the air flow into and out of the lungs.

Heart and lung transplant: if the lungs become badly damaged the only option my be to replace the damaged lungs.

Possible CF treatments for the future

In gene therapy the genotype and hence the phenotype of target cells is altered. This is done by:

- inserting normal alleles of the gene into the target cells (by using either genetically modified virus to infect the target or using liposomes
- the normal form of the gene is transcribed and translated
- a functioning protein is produced in the target cells

How genes are inserted using viruses:

- the DNA sequence that allows it to replicate is removed
- this is replaced with a normal allele of the desired gene
- when the virus infects the desired target the viral DNA becomes incorporated into the patients own DNA

How genes are inserted using liposomes:

- copy of the normal allele is inserted into a loop of DNA (plasmid)
- the plasmid is then combines with liposomes
- the positively charged head groups of the phospholipids combine with the DNA
- the liposomes fuse with the epithelial cell membranes and carry the DNA to cells

Testing for CF

Genetic testing:

The DNA is tested to see whether it contains the known base sequences for the most common mutations that cause cystic fibrosis.

How can genetic screening be used?:

- to confirm a diagnosis
- to identify carriers
- for testing embryos - amniocentesis involves inserting a needle into the amniotic fluid to collect cells that have fallen of the placenta and foetus
- chorionic villus sampling - a small sample of placental tissue is removed, either through the wall of the abdomen or through the vagina.
- testing before implantation - when carrying out in vitro fertilisation it is possible to test an embryo before it has implanted in the uterus. a cell can be removed from an embryo, the cells can then be analysed and used to decide whether to place the embryo into the womb.