genes and health Cheat Sheet

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Genetic screening

Amniocentesis	Chrionic Villus Sampling	NIPD (non-invasive prenatal diagnosis)	PGD (preimplantation genetic diagnosis)
• Done at 15-17 weeks of pregnancy	• Done at 8-12 weeks of pregnancy	• Done at 7-9 weeks of pregnancy	• N/A (for when it's done)
• A needle is placed into the amniotic fluid to collect foetal cells that have fallen off the placenta or foetus	• A small sample of the placenta is taken either through the wall of the abdomen or through the vagina	• DNA fragments are taken from the mother's blood plasma (most of it is cffDNA) for analysis	• Couple undergo IVF (30% success rate for women under 35)
• Allows the parents to make informed choices about future treatment, prepares parents for a child with a disease, gives the parents option for abortion.	• N/A	• Less traumatic procedure	• Only implants healthy embryos
• 1% risk of miscarriage	• Claimed 1-2% risk of miscarriage		• Procedure involving IVF can be regarded as unethical because the embryos are discarded, emotionally traumatic.
• Invasive method	**• Invasive method**	**• Non-invasive**	**• Non-invasive**

Processes for topic 2

Process of DNA replication	Process of transc-ription	Process of transl-ation

Processes for topic 2 (cont)

Semi-cons-ervative replication is where the DNA molecule unwinds and unzips because of the enzyme DNA helicase so the hydrogen bonds between the bases break. The free DNA nucleotides line up alongside each single DNA strand and hydrogen bonds form between the comple-mentary	The DNA helicse unzips and unwinds the DNA at the gene that codes for the desired protein. The enzyme RNA polymerase then attaches to the DNA. The template strand is transcribed - an mRNA molecule is created using compleme-ntary RNA nucleo-tides. Phosphodi-	The mRNA molecule attaches to a ribosome. The mRNA binds to the small subunit so that 2 mRNA codons face the 2 binding sites of the larger subunit. A tNRA anticodon sequence comple-mentary to the mRNA codon sequence brings along a particular

CF stages

Cystic Fibrosis (cont)

bases. The enzyme DNA polymerase links the adjacent nucleotides with phosphodi-ester bonds in conden-sation reactions to form new comple-mentary strands.

ester bonds form between the RNA nucleotides of the mRNA strand. The mRNA moves out of the nuclear pores of the nuclear envelope.

amino acid. Within the cytoplasm, free amino acids become attached to the correct tRNA molecules. Comple-mentary anticodon UAC hydrogen bonds to the start codon. The second codon faces the next binding site and the comple-mentary tRNA amino acid complex binds to it. A peptide bond forms between the 2 amino acids via a conden-sation reaction between the amine group of the amino acid and carboxyl group (COOH) of the next, forming a dipeptide. The tRNA returns to the

Remember this is for a healthy individual.	Remember this is for a CF sufferer.
1. Na+ is actively pumped across the basal membrane	1. Cl- is pumped into the cell across the basal membrane.
2. Na+ diffuses through sodium channels in the apical membrane	2. Cl- diffuses through the open CFTR channels.
3. Cl- diffuses down electrical gradient	3. Na+ diffuses down the electrical gradient into the mucus
4. Water is drawn out of cells by osmosis due to the high concentration in the fluid tissue	4. Elevated salt concen-tration in the mucus draws water out of the cell by osmosis.
5. Water is drawn out of the mucus by osmosis	5. Water is drawn into the cell by osmosis

Cystic Fibrosis		
Respir-atory system	**Reprod-uctive system**	**Digestive system**
The cilia in the lungs of a CF sufferer would struggle to beat/move mucus out of the lungs due to the mucus being abnormally thick and sticky (viscous). This would mean that there is an increased risk of lung infections.	**Males:** The sperm cannot leave the testes because the vas deferens is blocked by the highly viscous mucus. This means that the vas deferens is absent so sperm cannot pass through.	The pancreati enzymes cannot enter the intestines because the pancreati duct is blocked by the thick mucus. Due to digestion being les efficient, high energy diet is required. Pancreat enzymes that are trapped behind th thick mucus damage the pancreati cells (e.g ones that produce insulin) and cysts form in th pancreas as a result. Diabetes may also occur.

cytoplasm where it can collect another amino acid 9. This process continues until a stop codon is reached. There are no anticodons to thee codons so no amino acids. Polype- ptide chain detaches from the ribosome.

Cystic Fibrosis (cont)

Thicker mucus is produced because the chloride ions cannot leave the cell through the CFTR protein channel and sodium ions don't move out of the cells so water moves into cells by osmosis.

Females: A mucus plug develops in the cervix which stops the sperm from reaching the egg so female sufferers with CF are less likely to become pregnant and are more likely to be infertile.

The thickness of the mucus depends/varies on the mutation because different mutations will have different effects on the protein produced and the chloride ion transport is affected by the extent of changes to the CFTR protein.

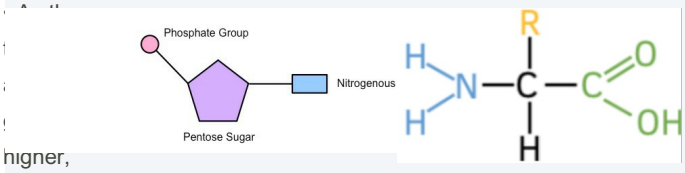
CP 4 - Enzymes

Variables	Controls (same)	Repeats (why?)	Results

CP 4 - Enzymes (cont)

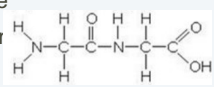
• IV: concentration of the enzyme	• Temperature (C)	• To compare optimum temperature for enzyme-substrate complex to form
• DV: time in seconds taken for the enzyme to break down substrate (using a stopclock)	• Volume of enzyme (cm3)	
	• Volume and concentration of the substrate	
	• pH	

Mononucleotide diagram



higher, the active site of the enzyme denatures faster.

Dipeptide diagram



You need to also draw water on. Remember, the O is on top and 2 Hs at the bottom.

Definition of a recessive allele

Recessive alleles are a different form of a gene and have the same two alleles (e.g. rr) and they are not expressed in the presence of a dominant allele. They are in the same locus of a chromosome but are a different form of a gene.

Definition of a gene

A gene is a sequence of bases on a DNA molecule that codes for a sequence of amino acids in a polypeptide chain

Definition of template

Where the RNA nucleotides attach for transcription

Definition of an allele

A different version of a gene found on a chromosome.

Structure of an amino acid

You need to know this for the exam since you have to draw it out.

Definition of a dominant allele

A dominant allele is the one that is expressed in the same phenotype (e.g. RR) and is found on the same locus of a chromosome.

Compare and constract diffusion + active transport

- both diffusion and active transport move molecules through the phospholipid bilayer/cell surface membrane
- molecules can move through proteins in both diffusion and active transport
- diffusion occurs **down** a concentration gradient whereas active transport occurs **against** a concentration gradient
- diffusion is a **passive process** that doesn't require ATP whereas active transport does require ATP

Compare and constract endocytosis + exocytosis

- **both endocytosis and exocytosis involve the usage of vesicles**
- both endocytosis and exocytosis **involve energy from ATP**

Compare and constract endocytosis + exocytosis (cont)

- **exocytosis** involes **molecules/substances leaving the cell** whereas **endocytosis** involves **substances/molecules entering the cell**
- **exocytosis** involves **vesicles fusing with the cell surface membrane** whereas **endocytosis** involves the **formation of vesicles** from the cell surface membrane

Human lung adaptations

- Rate of diffusion is proportional to surface area - alveoli have a large surface area
- Rate of diffusion is proportional to difference in concentration - breathing maintains a difference in gas concentrations
- Rate of diffusion is proportional to difference in concentration - blood flow maintains a difference in gas concentrations
- Rate of diffusion is inversely proportional to diffusion distance - walls of alveoli and capillaries are one cell thick
- Diffusion distance is reduced due to flattened cells forming alveoli and capillary walls
- Rate of diffusion is proportional to diffusion constant - cell membranes are relatively permeable to non-polar gas molecules

Human lung adaptations (cont)

Perfect exam answer (4-6 marks): The human lungs are adapted for gas exchange because the diffusion distance for gas exchange is reduced due to the flattened cells forming alveoli and capillary walls. The rate of diffusion is proportional to the surface area because the alveoli have a large surface area for gas exchange. Furthermore, the walls of the alveoli and capillaries are one cell thick so the rate of diffusion is inversely proportional to the diffusion distance and blood flow maintains a difference in gas concentrations.

NB: Fick's law states that the larger the surface area, difference in concentration and shorter the diffusion distance the quicker the rate. **You need to remember this for these kinds of exam questions.**

Sim + diff between fac. diffusion and AT

Similarity: Both facilitated diffusion and active transport use carrier/transport proteins

Difference: Facilitated diffusion is a passive process that doesn't require ATP whereas active transport uses ATP for energy.

CP 3 - Beetroot practical

Variables	Controls (how?)	Repeats (why?)
• IV: temperature of the water (Celsius)	• Volume of distilled water • Size of beetroot piece	• To calculate the permeability of the beetroot membrane
• DV: % transmission of light through resulting solution using a cuvette	• Time left in water	

Why is genetic screening used?

- Results confirm diagnosis
- for testing embryos
- to identify carriers

tRNA and mRNA

tRNA	mRNA
• folded/clover leafed	• straight folded chain
• size/length of tRNA is constant	• size/length of mRNA is variable
• tRNA has anticodons/amino acid binding sites	• mRNA has codons
• both contain RNA mononucleotides	
• both are single stranded	

Difference between transcription and replication

DNA Replication	Transcription
• Replication produces **double stranded DNA molecules**	• Transcription produces a **single stranded RNA molecule**
• Replication involves **DNA nucleotides**	• Transcription involves **RNA nucleotides**
• Replication uses **DNA polymerase**	• Transcription requires **RNA polymerase**

Difference between transcription and replication (cont)

• Replication produces **identical copies**	• Transcription produces a **complementary copy**

Globular and fibrous proteins

• Both globular and fibrous proteins are **chains of amino acids joined by peptide bonds**

• Both globular and fibrous proteins **contain hydrogen/disulfide/ionic bonds**

• Globular proteins have **hydrophillic groups** on the **outside** (soluble)	• Fibrous proteins have **hydrophobic groups** on the outside (insoluble)
• Globular proteins have **tertiary or quaternary structures**	• Fibrous proteins have **little to no tertiary structure**
• Globular proteins are **folded into compact shapes**	• Fibrous proteins have **long chains**

Compare and constrast deletion & substitution

• Deletion could affect every codon but substitution will **only affect one codon.**

Compare and constrast deletion & substitution (cont)

• Deletion is **more likely to affect the position** of the start codon/stop codon and results in a different sequence of amino acids whereas substitution may **not affect the sequence of amino acids.**

• Substitution **may code for the same amino acid** due to the **degenerate nature** of the genetic code.