Gene Expression
ANSWERS

DNA

Genotype and Phenotype
STOP AND CHECK (PAGE 7)

- **DNA:** The genetic code, made up of nucleotides in a double helix structure.
- **Chromosome:** A long molecule of DNA.
- **Gene:** A length of DNA that codes for a particular trait (e.g. eye colour).
- **Allele:** An alternative form of a gene (e.g. brown eyes or blue eyes).
- **Genotype:** The combination of alleles for a particular trait (one allele from each parent).
- **Phenotype:** The physical expression of the genotype, i.e. the trait exhibited.
- Proteins are big molecules that have special jobs in the body. A cell receives instructions from DNA and then makes the protein that forms the physical expression of a particular phenotype. Some proteins create actual structures in our bodies, and some create enzymes(biological catalysts which speed up reactions).

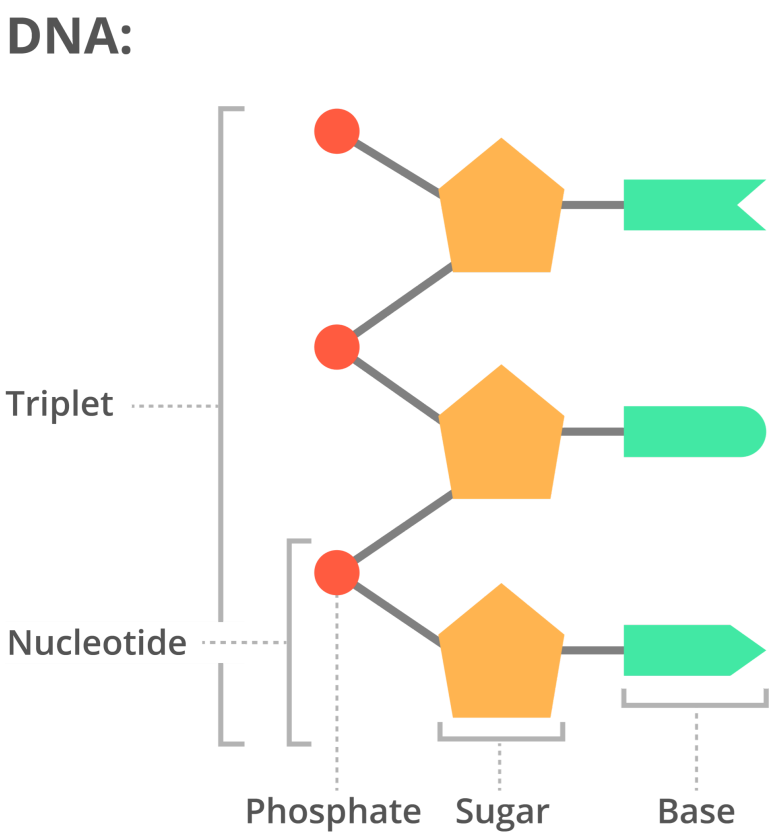
Structure of DNA
STOP AND CHECK (PAGE 9)

- DNA is shaped like a ladder twisted up, which makes it look like the shape of a double helix. The sides of the ladder are made up of sugar and phosphate, and the rungs of the ladder are made up of base pairs (A and T or C and G). A set of a sugar, a phosphate, and a base is called a nucleotide.
- Triplets are a series of three bases in DNA. Each triplet codes for a specific amino acid, which helps to provide the instructions to make a specific protein.

DNA

QUICK QUESTIONS (PAGE 9)

- The genotype of an individual is determined by which combination of alleles for a particular gene are inherited from each parent. For each gene, we receive one allele from our father and one allele from our mother, and this is what makes up our genotype. The phenotype of an individual is determined by the genotype, and is the physical expression of that genotype for a particular gene. For example, the genotype for eye colour might be one blue eye colour allele and one brown eye colour allele, and the phenotype, or trait expressed, would be brown eyes.
- DNA is shaped like a ladder twisted into a double helix shape. The sides of the ladder are made up of sugar and phosphate, and the 'rungs' of the ladder, in the centre of the structure, are made up of pairs of bases (A pairs with T, and C pairs with G). A set of a sugar, a phosphate, and a base is called a nucleotide.



Protein Synthesis

Protein and RNA

STOP AND CHECK (PAGE 12)

- Proteins are made in ribosomes, which are organelles in the cytoplasm of cells.
- RNA is only single-stranded, so half a ladder, as opposed to double-stranded like DNA. This is because it only carries the instructions for the protein production of one single gene, whereas DNA carries all of the instructions that code for every single protein. The DNA remains in the nucleus of the cell, whereas the RNA carries the single copy of the gene to a ribosome in the cytoplasm of the cell for protein production. Also, whenever a strand of DNA has a T base, it is replaced with a U base when DNA is copied (transcribed) into the RNA.
- **Transcription:** the process of making RNA from DNA, the first step of protein synthesis. Translation: the process of making a protein from RNA, the second step of protein synthesis.

Transcription

STOP AND CHECK (PAGE 14)

- Transcription occurs in the nucleus of the cell.
- During transcription, an enzyme unzips the DNA and opens the required gene. It
  breaks the hydrogen bonds between base pairs separating them and splits the DNA ladder in half. The enzyme then uses the template strand to create RNA, adding complementary nucleotides, so that it is now identical to the coding strand. The RNA then breaks off from the template strand, the DNA zips back up again, and the RNA leaves the nucleus through the nuclear pore in the nuclear membrane.
- RNA is a single-stranded copy of a gene that carries the necessary genetic code from the nucleus of the cell to a ribosome so that the protein for that gene can be made. It is made by an enzyme that splits a particular gene on

DNA into two strands and then copies one strand and adds the complementary nucleotides, creating an RNA strand.

Translation

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- Translation occurs when the mRNA travels to a ribosome. The ribosome binds to the START codon of the mRNA strand, and then the tRNA containing the complementary anticodon enters the ribosome, bringing with it the correct amino acid which the particular codon codes for. The ribosome then moves to the next codon of the mRNA strand and repeats the process, and bonds the new amino acid to the previous one with a peptide bond. By the time the ribosome reaches the STOP codon, there is a fully-formed polypeptide chain of amino acids, which folds up into different structures with specific functions and forms the primary structure of a protein.
- Amino acids are made up of the codes given in triplets of bases in DNA, and therefore subsequent codons in mRNA. A specific sequence of amino acids makes up a specific protein.

Connecting it Together

STOP AND CHECK (PAGE 20)

- The base sequence in a single gene is organised into triplets of bases, which transcribe into codons when the string of mRNA. The sequence of codons on the mRNA determines the type of amino acids made in the polypeptide chain that the ribosome creates, which forms the protein. The sequence of amino acids on the polypeptide chain determine the structure and the function of the protein formed.
- The special enzyme creates mRNA with bases that are complementary to the template strand of DNA so that the final mRNA strand is identical to that of the coding strand of the transcribed DNA. Also, during translation, in order for the tRNA to successfully attach itself to the mRNA, it must contain an anticodon on one end which is complementary to the START codon of the mRNA.

Protein Synthesis

STOP AND CHECK (PAGE 20)

- DNA holds all of the necessary genetic information for proteins, and therefore all body functions and traits, to be made. RNA is responsible for carrying the information from the DNA in the cell's nucleus to the ribosomes in the cell's cytoplasm so that the necessary protein that the DNA codes for can be made. RNA is made by a special enzyme that unzips the DNA in the nucleus to access the necessary gene, splits the DNA into two strands, and then uses one strand, the template strand, to create a string of RNA with bases that are complementary to those on the template strand. The string of RNA, therefore, becomes identical to the other strand of DNA, the coding strand.
- Although transcription and translation are separate processes, they work together consecutively. Translation happens only after transcription has taken place, and both processes are necessary in order for DNA to be made into a protein and therefore expressed in the body's function. The transcription process occurs in the cell's nucleus, whereas translation occurs in the cell's cytoplasm. Transcription involves a special enzyme splitting DNA in half, into two strands, so that one strand can be used as a template to create mRNA. Once mRNA is formed and has travelled from the nucleus to a ribosome in the cytoplasm, only then can the process of translation take place. Translation doesn't require the same special enzyme as transcription. Instead, the ribosome and matching tRNA are responsible for creating the particular sequence of amino acids that the mRNA codon sequence codes for, therefore making a protein that will carry out a specific function.
- The information for every single protein is encoded within the DNA of organisms. Each cell contains the entire genetic code. However, the cells only undergo transcription of relevant genes to their function. So, while each cell contains the entire genetic code, it will only access the genes that are necessary to its function.

Mutations

Defining Mutations

STOP AND CHECK (PAGE 21)

- Mutations are permanent changes in the base sequences of DNA.
- The main causes of mutations are an accidental change when the cell is replicating its DNA and by mutagens such as radiation, chemicals, or viruses.

Effect of Mutations

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- When a mutation occurs, the sequence of triplets in the DNA changes, which causes the sequence of codons in the mRNA to change during transcription, which causes a different sequence of tRNAs to come into the ribosome during translation, which causes a different sequence of amino acids in the polypeptide chain, which causes the protein formed to fold up into a different structure, which means that the protein might not function the way it is supposed to, or at all.

Types of Mutations

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- A substitution mutation results in a whole new amino acid being added to the polypeptide chain, and insertion and deletion mutations cause a frameshift in the whole sequence of amino acids because each base is shuffled into a new codon.
- Frameshift is when every triplet of bases from the mutation onwards is changed because every base is shuffled into a new codon. This creates a totally new set of amino acids, which therefore completely changes the sequence on the polypeptide chain. This means that the protein formed is highly unlikely to be able to function because it is so different to how it is supposed to be.

Metabolic Pathways

Enzymes

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- Enzymes allow all biological reactions to take place. Substrates attach themselves to the active site of enzymes, the enzymes work on the substrates and then release them.
- Enzymes are specific for particular substrates because only specific substrates can fit into each enzyme's active site.

Metabolic Pathways

STOP AND CHECK (PAGE 26)

- A metabolic pathway is a series of enzyme-catalysed reactions, in which the product of each enzyme reaction is the reactant for the next, and so on, until the final product is made.
- The series of reactions in a metabolic pathway is created by the product of each enzyme reaction becoming the substrate for the next enzyme's reaction, and so on. Therefore, there may be multiple different enzymes involved in each metabolic pathway because each time there is a new substrate, there needs to be a specific enzyme that the substrate fits.

Mutations in Metabolic Pathways

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- If just one enzyme is not produced or is mutated and non-functional, it won't catalyse the necessary reaction to create a product, which means that there will be no substrate for the next enzyme in the metabolic pathway, which means that this next enzyme cannot catalyse the next reaction, which leads to the final product for the metabolic pathway not being formed. Since the final

product isn't formed, the body could be missing a vital substance, which could cause disease. Also, the enzymes before the mutated enzyme on the metabolic pathway will still be catalysing reactions, creating a build-up of substrates that aren't being used.
- A build-up of substrates can be toxic, which can cause disease.

Mutations in Metabolic Pathways

QUICK QUESTIONS (PAGE 29)

- Tyrosine hydroxylase would attach itself to the tyrosine substrate with its active site, which would create a reaction, which would form DOPA as its product. DOPA, now acting as the substrate, would then attach itself to L-amino acid decarboxylase, causing a reaction, and forming dopamine as the final product.
- A metabolic pathway is a series of enzyme-catalysed reactions, in which the product of each enzyme reaction is the reactant for the next, and so on until the final product is made. This is seen here, tyrosine is the reactant for the enzyme tyrosine hydroxylase, and the product of this reaction (DOPA) becomes the reactant for the next enzyme (L-amino acid decarboxylase), and the product of this reaction produces dopamine as the final product.
- If there was a substitution mutation in either of the enzymes, the amino acid affected may or may not change, depending on which bases make up the codon of the amino acid. If the amino acid does not change, this will cause no change in the sequence on the polypeptide chain of the enzyme, and therefore the structure and function of the enzyme will not be affected. However, if the amino acid does change, this will cause a slight change in the sequence on the polypeptide chain of the enzyme, meaning that its structure might change. If the structure changes, there is a chance that the necessary substrate (tyrosine or DOPA) won't be able to fit into the active site of the enzyme anymore, which would cause the metabolic pathway to cut short, and therefore not produce dopamine. If there was an insertion or deletion mutation in either the tyrosine hydroxylase or the L-amino acid decarboxylase, it is much more likely that the whole amino acid sequence of the enzyme will change, which is likely to cause the enzyme to stop functioning. If this is the case, the metabolic pathway will also be cut short because the

enzyme-catalysed reactions cannot take place, and dopamine will not be produced.

Environment and Phenotype

Environment and Phenotype

STOP AND CHECK (PAGE 31)

- One example where environmental factors might alter the physical expression of a trait is nutrition. If a child does not receive all the necessary nutrients at a young age, this is likely to affect their body's ability to grow to its full potential, therefore affecting the trait of height.

Environment and Phenotype

QUICK QUESTIONS (PAGE 31)

- A person's DNA determines the limit, or extent, to which a particular trait will be physically expressed, and then environmental factors have influence over the expression of that trait within the limit that the DNA provides. For example, a person's DNA will determine their full potential of how tall they could possibly grow. However, they may not necessarily grow to this maximum height, depending on environmental factors, such as their nutrition as a child, or any injuries or surgeries they may endure when they are not fully grown.