Year 11 Human Biological Sciences (Stage 2)

Extended Response 4

Complete the following extended response in a standard answer booklet.

*Spinal motor atrophy (SMA) is a genetic disease characterised by weakness and muscle wasting. It is caused by a genetic error that damages the nerves that control most of the body’s muscles. As the nerves degenerate, the muscles grow weaker because they are not being stimulated.*

1. Describe the structure of DNA. [4]
2. Explain how an error in the DNA code can cause a disease like SMA. Describe the basic steps of protein synthesis in your answer. [8]
3. SMA is an autosomal recessive disorder. Describe the most likely way this disease would be passed from parents to child. Explain the meaning of, and use each of the following terms in your answer.

* Heterozygous and homozygous
* Recessive and dominant
* Genotype and phenotype

[8]

1. [Any 4 marks]

* Double helix
* DNA strand is coiled around histones/structural proteins
* Alternating sugar and phosphate backbone
* Combinations of four bases form cross bridges
* Complementary base pairing / A-T, G-C

1. [Any 8 marks]

* DNA molecule unzips (at location of gene)
* Messenger RNA is formed by copying the gene code
* mRNA is complementary to the template DNA code except that T is replaced with U
* mRNA leaves the nucleus and travels to a ribosome
* transfer RNA brings amino acids from the cytoplasm
* ribosomes read the mRNA code and then link the amino acids from the tRNA into a protein.
* If the DNA code is incorrect, then the mRNA code will be wrong
* Ribosomes will assemble amino acids in the wrong order
* The resulting protein may not function properly

1. [Any 8 marks]

* Most likely inherited from two heterozygous parents who have a normal allele and an SMA allele
* The genotype of the parents would be Ss
* The parents do not have SMA because they have a normal allele which is dominant
* Dominant alleles mask recessive alleles
* Each parent passes one allele down to their child
* Therefore the child is homozygous (two alleles the same)
* The child’s genotype is aa
* Because both alleles are recessive the child shows the disease
* Phenotype is the presence of absence of the disease
* The phenotype of the parents is normal; the phenotype of the child is SMA