

AS 90715 Describe the role of DNA in relation to gene expression

Mutations

(2010: 2b)

(b) A single **substitution** mutation in the DNA **may or may not** lead to a change in the functional protein. Discuss the effect a single substitution mutation in the DNA may have on the formation of a functional protein.

You must include the example from part (a) above in your explanation.

You should include in your answer:

- how a mutation would be **passed on** during protein synthesis
- **redundancy** of the genetic code
- the **position** of the mutation in the DNA strand.

(2009:1d)

People with mutations in the NF1 gene develop neurofibromatosis type 1, a disease of the nervous system that affects 1 in 3 500 people worldwide. Several different mutations result in neurofibromatosis. Some of these mutations involve the RNA transcript.

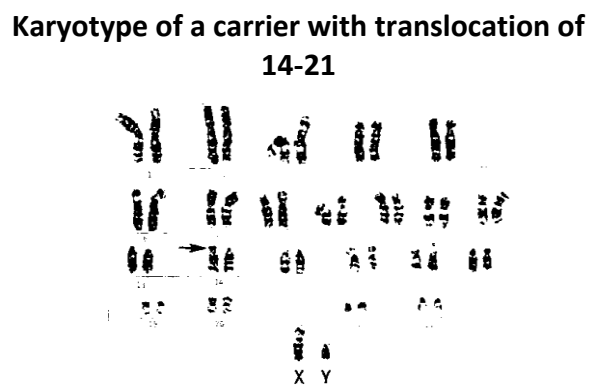
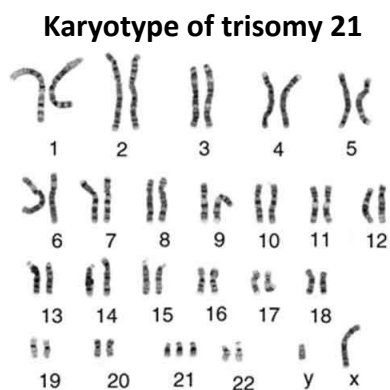
Discuss how TWO types of mutation may arise in the NF1 gene, **and** the effect these mutations could have on the control of cell division.

In your answer you should:

- describe each type of mutation
- explain the relationship between the size of the NF1 gene and the occurrence of mutations
- explain the effect each mutation may have on the control of cell division.

(2009:3)

Down syndrome, or trisomy 21, is due to the presence of all or part of an extra 21st chromosome. People with trisomy 21 have impaired cognitive ability, impaired physical growth and a characteristic appearance. Nearly all cases of Down syndrome result from non-disjunction of chromosome 21 during meiosis. However, a few cases can arise from a translocation mutation. In this situation, part or all of chromosome 21 is fused to another chromosome, usually chromosome 14.



An individual with a translocation 14-21 can be phenotypically normal, and is called a *carrier*.

- (a) Explain why a carrier can be phenotypically normal.
- (b) Discuss the genetic differences between individuals with Down syndrome due to trisomy 21, and individuals with Down syndrome due to a translocation mutation.

In your answer consider:

- the chromosome complement of individuals affected by each type of mutation
- the process of meiosis
- the genotypes of parents with affected children.

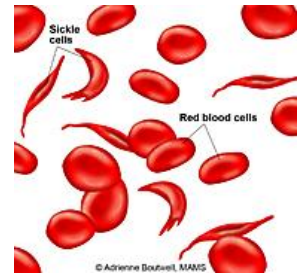
(2007:2)

Human haemoglobin is formed from two polypeptides, the α -globin and β -globin chains. Glutamic acid is normally the sixth amino acid in the β -globin chain.
 The first part of the DNA template sequence, for amino acids one to eight, of the normal β -globin chain is:

CAC GTG GAC TGA GGA CTC CTC TTC

(a) In the space below, give the **sequence** of the corresponding **mRNA** strand, which will code for these amino acids.

First Position	Second Position			Third Position	
	U	C	A	G	
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	Stop	Stop	A
	Leu	Ser	Stop	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G



People with sickle-cell anaemia have a different base sequence that codes for valine instead of glutamic acid as the sixth amino acid.
 The mRNA codons for glutamic acid are GAA and GAG, and the codons for valine are GUU, GUC, GUA and GUG.

- (b) Describe the changes to **both** the mRNA codon and the DNA base sequence as a result of this mutation.
- (c) Discuss the **effect** of this mutation on the **red blood cell**, through the mutation's effect on:
- mRNA
 - the amino acid sequence
 - the general structure of the protein
 - the function of the protein
 - the shape of the red blood cell.

(2006:3)

Genes are found on chromosomes, and eukaryote cells usually contain two copies of each chromosome. The members of a pair of chromosomes have the same sequence of genes, but the genes may come in slightly different forms.

- (a) On the diagram below, show the position of alleles for an individual who is:
- homozygous dominant for gene A
 - heterozygous for gene B
 - homozygous recessive for gene R.



People affected by Duchenne muscular dystrophy (DMD) lose muscle function from an early age and rarely survive to adulthood. The disease is caused by a mutation in a gene found on the short arm of the human X chromosome. The dominant allele (M) results in normal muscle function, while the recessive allele (m) produces the slow, irreversible muscle wasting that is characteristic of this disease.

- (b) Give ALL the possible genotypes for the phenotypes listed below:
- female not affected
 - female affected
 - male affected
 - male not affected
- (c) (i) A female who is heterozygous for the DMD allele has children with an affected male. Use a Punnett square to show all possible genotypes for the offspring of this couple.
- (ii) From your Punnett square, list the phenotypic and genotypic proportions for male and for female offspring.

Phenotypic proportions
male:
female:

Genotypic proportions
male:
female:

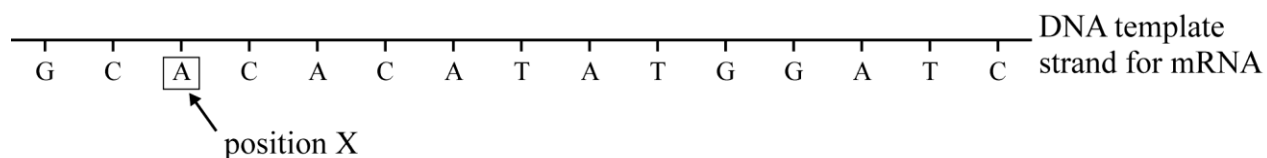
- (d) Explain why more males than females are affected by Duchenne muscular dystrophy.

Many metabolic pathways are controlled by multiple genes. An example is the metabolic pathway that produces normal skin pigmentation. Albinism, which is the total lack of pigment, can be caused by a mutation in any one of the genes controlling this pathway.

- (e) Discuss the fact that it is possible for two albino parents to have a child with normal skin pigmentation.

(2005:2)

The base sequence from the end of a gene is shown below. Use it and the table given to answer the questions that follow.



- (a) Use the table to determine the sequence of amino acids formed from this portion of the gene.
- (b) The table is in the form of codons of mRNA. Codons are made up of nucleotides. Describe what makes up the nucleotides of **codons**.
- (c) Discuss why a substitution point mutation at the position marked X will have very little effect on the polypeptide formed, whereas a deletion point mutation at position X would prevent production of the same protein.

First Position	Second Position			Third Position
	U	C	A	G
U	Phe	Ser	Tyr	Cys
	Phe	Ser	Tyr	Cys
	Leu	Ser	Stop	Stop
	Leu	Ser	Stop	Trp
C	Leu	Pro	His	Arg
	Leu	Pro	His	Arg
	Leu	Pro	Gln	Arg
	Leu	Pro	Gln	Arg
A	Ile	Thr	Asn	Ser
	Ile	Thr	Asn	Ser
	Ile	Thr	Lys	Arg
	Met	Thr	Lys	Arg
G	Val	Ala	Asp	Gly
	Val	Ala	Asp	Gly
	Val	Ala	Glu	Gly
	Val	Ala	Glu	Gly