2018 Mutations MCQ

2018 / H2 / AJC PRELIM / P1 Q15

1 Tay-Sachs disease is a fatal neurodegenerative disease which is caused by a mutation in the hexosaminidase A (Hex A) gene located on chromosome 15.

Part of the sequence of the non-template (coding) DNA strand of the normal Hex A allele and the mutated Tay-Sachs allele are shown below. The sequences are the same as the mRNA sequence of both alleles.

DNA sequences of normal Hex A allele:

Amino acid position		424	425	426	427	428	429	430	431	
Non-template DNA	5’ …	CG T	AT A	TC C	TAT	GG C	CCT	GA C	TGT	…3’

DNA sequences of mutated Tay-Sachs allele:

Amino acid position		424	425	426	427	428	429	430	431	
Non-template DNA	5’ …	CG T	AT A	TCT	AT C	CTA	TG G	CC C	TG A	…3’

For both alleles, 9 different amino acids are encoded for by the DNA triplets:

Amino acid	DNA triplet
Arg	CGT
Asp	GAC
Cys	TGG, TGT
Gly	GGC
Ile	ATA, ATC

Amino acid	DNA triplet
Leu	CTA
Pro	CCC, CCT
Ser	TCC, TCT
Tyr	TAT
Stop codon	TAG, TAA, TGA

Which statement is true?

A The disease is caused by the deletion of one DNA nucleotide.

B The Hex A protein encoded for by the Tay-Sachs allele is non-functional due to a frameshift mutation.

C The polypeptide encoded for by the Tay-Sachs allele has the same number of amino acids as that encoded by the normal Hex A allele.

D At amino acid position 431, there is a silent mutation.

2018 / H2 / AJC PRELIM / P1 Q16

2 Edwards’ syndrome is a common autosomal disorder caused by chromosome aberration.

- Edwards’ syndrome is a trisomy of chromosome 18 that affects all cells.
- The extra chromosome is most often of maternal origin.

- In approximately more than 50% of individuals diagnosed with the syndrome, two of the three chromosomes 18 present are found to be nearly genetically identical.

What can be correctly concluded from this information?

A Non-disjunction of chromosome 18 commonly occurs during gamete formation in females.

B Non-disjunction of chromosome 18 occurs more frequently during meiosis I than in meiosis II.

C Non-disjunction of chromosome 18 occurs most commonly in female embryos.

D Two of the three chromosomes 18 are nearly genetically identical due to uneven crossing over resulting in chromosomal translocation.

2018 / H2 / EJC PRELIM / P1 Q5

3 The figures below show the complete karyotypes of 2 rodents of the same species. In this species of rodents, males are heterogametic.

Rodent A (Male)

Rodent B (Female)

Which of the following observations is not true?

A A chromosomal aberration occurred in the ovary of the mother of rodent **B**.

B Rodent **A** is diploid and 2n = 16.

C Rodent **B** has 1 missing X chromosome.

D Non-disjunction of autosomes occurred in rodent **B**.

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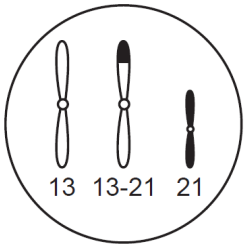
4 Down's syndrome can be caused by a trisomy of chromosome 21, but can also result from translocation of chromosome 21 onto chromosome 13, forming a single chromosome 13-21.

The diagram shows chromosomes 13 and 21 in the nucleus of a diploid (2n) testis cell from a phenotypically normal male carrier of a 13-21 translocation. This cell has a chromosome number of 45.



Which is **not** a likely outcome of fertilisation of normal oocytes by sperm from this male?

	chromosomes in sperm	embryo
A	13 and 21	2n = 46 normal phenotype
B	13-21	2n = 45 normal phenotype
C	13-21 and 21	2n = 46 Down's syndrome
D	13-21 and 21	2n = 47 Down's syndrome

2018 / H2 / MJC PRELIM / P1 Q13

QUESTION 5
Which statement(s) describe(s) how a gene mutation can lead to the production of a non-functional protein?

1 During transcription an incorrect nucleotide is added to a DNA molecule.

2 A codon in the mRNA transcribed from the mutated gene is changed.

3 The order of the bases in an anticodon on tRNA is altered during translation.

4 The sequence of nucleotides in the promoter of the gene is altered

A 2 only **B** 1 and 2 only **C** 2 and 4 only **D** 2, 3 and 4 only

2018 / H2 / RI PRELIM / P1 Q15

6. A geneticist determines that a particular human disease is caused by a gene mutation. The mutant allele contains a substitution of cytosine to adenine at position 334. The DNA sequence for bases 301 to 351 from the non-template strand of the normal allele is shown.

5'- ATG TTA CGA GGT ATC ATA CGA ACG GAG CGC GAA CTA GTT ACT CCC ATA AAA - 3'

Which of the following statements best describes a consequence of this mutation?

A A nonsense mutation has occurred resulting in no protein product being formed.

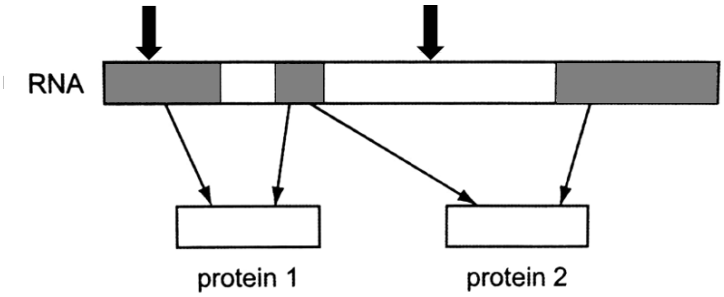
B The mutant protein contains fewer amino acids than the normal protein.

C A missense mutation has occurred resulting in a non-functional protein.

D There is no change in length of amino acid sequence due to the mutation occurring outside of the coding region.

2018 / H2 / RVHS PRELIM / P1 Q13

7 The diagram shows alternative splicing, in which the same mRNA can be translated to give two different proteins.



If a base-pair addition occurred at the DNA corresponding to the two sites indicated by arrows, what is the likely result on proteins 1 and 2?

	protein 1	protein 2
A	functional	functional
B	functional	non-functional
C	non-functional	functional
D	non-functional	non-functional

2018 / H2 / RVHS PRELIM / P1 Q14

8 The following shows a target sequence of interest.

5’ CGA GCT TTT ATA GAT TAT AGG CCT AAC AGA CTA 3’

3’ GCT CGA AAA TAT CTA ATA TCC GGA TTG TCT GAT 5’

The sequence can be digested by two different restriction enzymes. The sequences recognised by the restriction enzymes and points of action (indicated by *) are shown.

*Alu*I

5’ … A G * C T … 3’

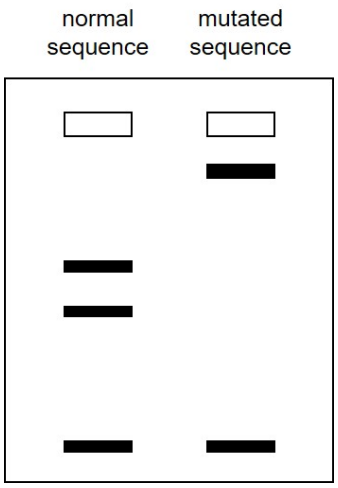
3’ … T C * G A … 5’

*Hae*III

5’ … G G * C C … 3’

3’ … C C * G G … 5’

A sample of the target sequence was digested with both restriction enzymes. The restriction fragments were then subject to gel electrophoresis. The same procedure was performed for a mutated target sequence.



Which of the following shows the mutation in the mutated target sequence?

	restriction site	type of mutation
A	*Alu*I	base-pair substitution
B	*Alu*I	inversion of restriction sequence
C	*Hae*III	base-pair substitution
D	*Hae*III	inversion of restriction sequence

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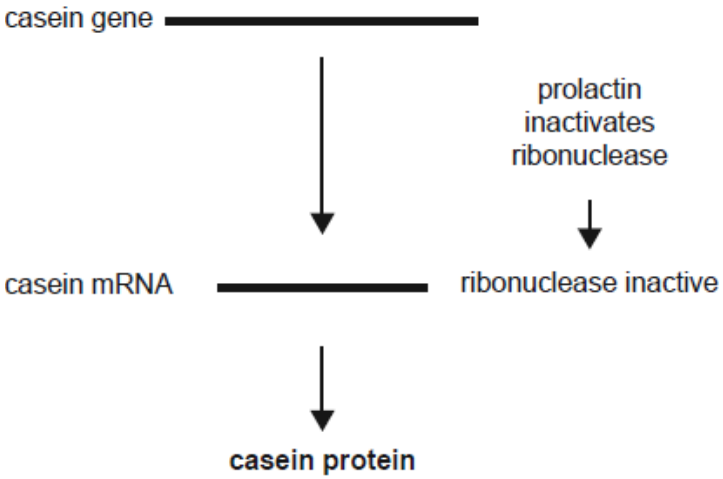
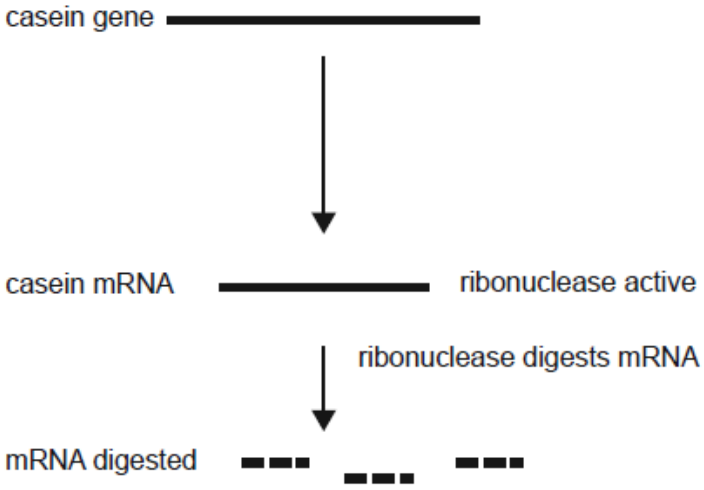
9 Casein is a major protein found in mammalian milk.

When the mammals are producing milk, the pathway for the production of casein can be represented as shown in the diagram below.



When the mammals are not producing milk, the pathway can be represented as shown in the diagram below.



Which one of the following conclusions can be made from the information above?

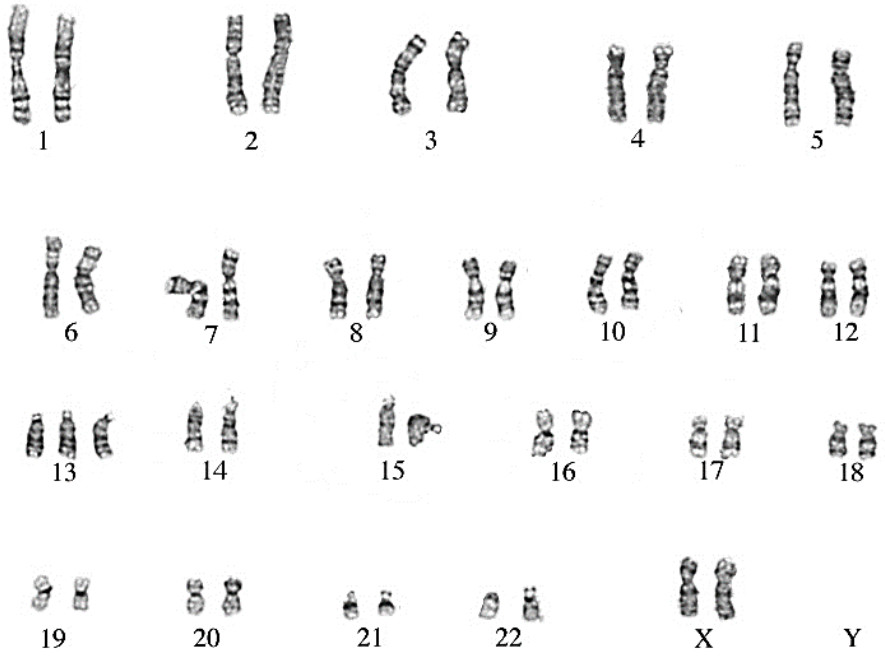
A Ribonuclease has the effect of turning on the casein gene.

B Casein is a repressor protein for milk production in mammals.

C The hormone prolactin allows for the expression of the casein gene.

D Mammals produce milk only in the absence of the hormone prolactin.

A newborn baby was diagnosed with Patau syndrome. The diagram below shows her chromosomes.



This is an example of

A frameshift mutation

B silent mutation

C aneuploidy

D polyploidy

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2018 Mutations MCQ ANS			
Question	**Answer**	**Question**	**Answer**
1	**B**		
2	**A**		
3	**D**		
4	**D**		
5	**A**		
6	**C**		
7	**C**		
8	**C**		
9	**C**		