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SBI 4U0 Genetics Unit Test Outline

Part A – Multiple choice - Knowledge - 25 marks:

DNA history (Scientists), structure of DNA and RNA, Role of enzymes in DNA replication and Protein synthesis, Operons, General knowledge of Chapter 7 techniques.

Part B – Multiple Choice – Thinking – 10 marks:

Questions about Chapter 7 techniques – Interpreting Results

Part C – The Big Question – 25 marks (Communication 10 marks, Application 15 marks):

Prepare (ahead of time) a cohesive answer with diagrams and explanations of terms (on big ledger paper).

The Big Question (25 marks)

Complete this question on the ledger paper provided. Be sure to use diagrams as part of your answer.

Question: Proteins go through complicated processes to become functional. There are many steps where mistakes can happen. Using a specific DNA template sequence, **EXPLAIN HOW ONE DNA MUTATION CAN CHANGE THE NORMAL TRANSCRIPTION/TRANSLATION/PACKAGING PROCESS AND CAN RESULT IN A NON-FUNCTIONAL PROTEIN.**

Criteria:

- It is easier to get full marks if you use all the terms but not necessary. You may discuss other terms as well.
- You must use diagrams in your answer to receive full marks.
- Explain normal protein synthesis and then how one mutation would change the process(es).
- Create a specific DNA template sequence to illustrate your concepts. A codon chart will be provided.
- It is vital that you connect your ideas together.
- Your answer must fit on one sheet of ledger paper, but you may use both sides.

Terms List:

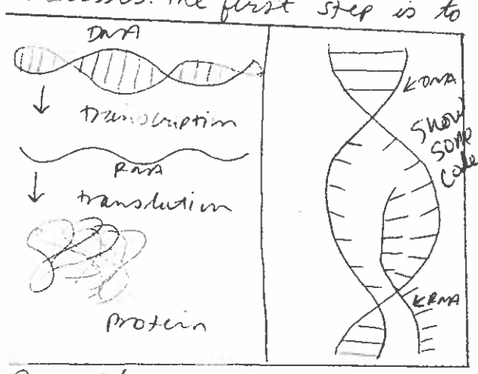
5'cap	mRNA	RNA polymerase
Anticodon	Missense mutation	RNA splicing
A-site	Non-coding strand	ribosomal subunits
ATP	Nonsense mutation	Silent mutation
Codon	P-site	Spliceosome
coding strand	Peptide linkage	Start codon
E-site	Point mutation	Stop codon
Exon	Poly A tail	Template
Genetic code	Primary mRNA transcript	Terminator
Intron	Promotor	tRNA

K 19/25 T 6/10 A 12/15 C 9/10 Total 46/60

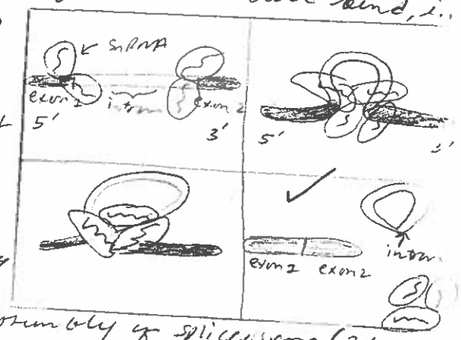
Ojala Khusal
ID: A6

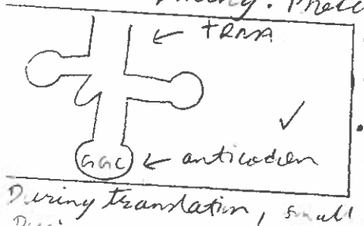
modifications + folding of this?

- In order to become functional, proteins must go through complicated processes. The first step is to synthesize mRNA from DNA. mRNA is used as a template that determines the amino acid sequence of the protein that it codes for. The double stranded DNA unwinds and unzips & for each gene only one strand of DNA is transcribed. This strand is called non-coding strand or a template strand. The other strand that is not transcribed is called a coding strand. During this process, an enzyme called RNA Polymerase catalyzes the synthesis of RNA



Promoter, a region on the DNA, indicate where RNA polymerase should bind, in order to start transcription. Terminators serve as a signal to stop transcription & the RNA detaches from DNA. After the mRNA is synthesized, it is modified by adding 5' cap (G nucleotide) to 5' end & 3' poly-A-tail (A nucleotides) to 3' end of primary mRNA transcript (Pre-mRNA). Non-coding region called introns, that are interspersed among coding region called exons, are removed in a process called splicing. Protein & RNA splicing involves assembly of spliceosome (2+ snRNA)





- tRNA links the codons on mRNA to the corresponding amino acid for protein synthesis. tRNA also contains anticodon that is complementary to specific mRNA codon.
- Ribosomal subunits provide a place for mRNA & tRNA to carry out protein synthesis. Protein synthesis is also called translation & it requires a lot of energy/ATP.

During translation, small ribosomal-subunit attaches to mRNA near start codon. During protein synthesis, tRNA binds to 3 sites: P(eptide) site, A(mino acid) site & E(xit) site. P-site: contains tRNA with growing polypeptide, A site: contain tRNA with next amino acid added to polypeptide chain, E side: uncharged tRNA is ejected. A peptide bond forms between first & second amino acid and so on, that holds the amino acids together. During termination mRNA reaches stop codon. The polypeptide is separated & released from the machinary & the polypeptide folds into 3-D shape & turns into protein readily to carry out cellular activities. The order of the codons of the mRNA determines the order of amino acid & growing polypeptide strand until eventually turns into complete protein.

DNA Sequence : TAC TCG CTG GGG AAG CCC GAC AAT ACT
RNA sequence : AUG AGC GAC CCC* UUC GGG CUG UUA UGA
Amino acid : met - Ser - ASP - PRO - Phe - Gly - Leu - Leu - Stop

- During replication, errors are very common, although enzymes do repair most of the errors, some result in mutation. Mutation causes change in the genetic code & that code is passed into daughter cells. Some mutation are harmful, some are beneficial & some are harmless.
- Some of the mutations include:
  - Point mutation: single base pair mutation (harmful)
  - Silent mutation: does not change Amino acid sequence (not harmful)
  - ↳ missence mutation: changes Amino acid sequence (harmful)
  - ↳ nonsense mutation: shortens a protein by introducing a stop codon (harmful)
- If there is a point mutation in the above sequence (Insertion/deletion) it results in frameshift mutation & missense mutation as it changes the amino acid sequence.
- If 12th base from above sequence is deleted the following will result

RNA sequence : AUG AGC GAC CCU UCG GGC UGU UAU GA
Amino acid : met - ser - ASP - Pro - Ser* - Gly - Cys - Tyr

Although the 4th Amino acid is same as the above / amino acids following that are different as a result of frameshift. The mutated code would be transcribed and undergo all the process of protein synthesis to synthesize a different protein than what DNA originally coded for. For example: If the genetic code produced by DNA was originally for black hair, but because of mutation & different amino acid sequence, it would synthesize protein that codes for blonde hair.

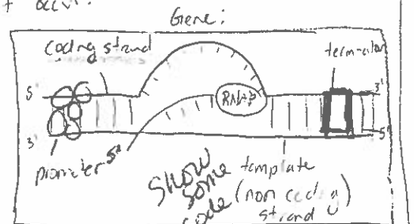
how is the 3D structure of the protein affected by changes in the amino acid sequence?

Genetic Code - a set of rules that determine how genetic information is made up through nucleotide sequences converted into amino acid sequence & then a protein in living cells

DNA - a double helix made up of complementary nucleotide base pairs bonded by hydrogen bonds
A=T C≡G

↳ in order to create proteins, protein synthesis must occur!

Transcription



1) Initation
- RNA polymerase binds to the promoter region known as T.A T.F box
  ↳ 25 nucleotides upstream
  ↳ made of a TA sequence that is easy to break due to 2 hydrogen bonds (G≡C has 3 H-bonds)
- double helix unwinds to be transcribed

2) Elongation
- RNAP builds in the 5' to 3' direction reading from the 3' to 5' template (non coding) strand

RNAP unwinds template to be transcribed ahead & behind DNA rebonds
- mRNA is complementary to template strand, however uracil pairs with adenine instead of thymine pairing with adenine

Termination
RNAP reaches the terminator which causes RNAP & mRNA that's been built to dissassociate from template strand

Modifications
↳ cells contain nucleases that chomp on DNA & RNA
↳ when the primary mRNA transcript leaves the nucleus & enters cytoplasm, it must be protected!

5' Cap - methyl group added to 5' end to protect from digestion & initate translation

Poly A Tail - 40 - 250 adenine sequence added to 3' end for protection from degradation

↓

Conversion to Mature mRNA

Introns = non coding (junk) regions that are not expressed

Exons = coding regions that are expresse

In order to remove these introns from the mRNA, a technique called RNA splicing occurs
↳ spliceosome - cuts out the introns from sequence & pins the remaining exons together

↓

Translation

↳ mature mRNA is transported to ribosome

↳ mRNA is read in multiples of 3 nucleotides which is a codon

tRNA

↳ recognizes the mRNA codon

Ribosome Structure:

made up of proteins & rRNA

↳ made up of 2 ribosomal subunits (large & small)
- sub units wrap around mRNA

(1) Initation

↳ Finder sequence in large sub unit is AUG & anticodon in small subunit is UCCU, this is what holds the ribosomal sub units together

- The start codon (AUG) is the initation codon for all proteins
- Attaches using UAC with MET amino acid attached at the P-site

A-site - tRNA carrying its corresponding amino acid (name's end) awaits for it to bind with growing chain

*P-site - where the polypeptide chain is held, tRNA from A site moves to P site

E-site - site where tRNA dissassociates & where & mRNA disperse

(2) Elongation
- New tRNA binds to A-site with corresponding amino acid
- peptide bond occurs between new amino acid in A with P site
- give the polypeptide chain attaches to amino acid the tRNA moves from A-site to P site & the previous tRNA with polypeptide goes to E-site & dissociates
- process keep occuring

Termination
- In order for the chain to stop going, the introduction of a stop codon such as UGA, UAA, etc must occur
- this will cause the ribosome, mRNA, & polypeptide to disperse

↓

Post Translation
- undergoes folding, phosphorylation ✓ quarternary step, etc

↓

Mutations

Substation

Silent mutation - occurs when one nucleotide is wrong but still codes for same amino acid ex. UAU & UA = tyr = tyr

missense mutation - occurs when one nucleotide is substituted & changes the amino acid ex. GAU & GA = Asp = G

nonsense mutation - occurs when one nucleotide is substituted & causes the sequence to stop ex UGC & UG = Cys = ST

Point Mutation = a disorder of the codon sequence which can cause severe effects
- through addition of a nucleotide which shifts nucleotides 1 to the right
- through deletion of a nucleotide which shifts nucleotides 1 to the left

DNA sequence = TAC CAC GGG TGA TTA GCT

mRNA sequence = AUG GUG CCC ACU AAU CGA

amino acid sequence = Met / VAL / PRO / thr / ASN / Arg

} original sequence (18 nucleotides)

* Now if there was an addition of nucleotide it would cause a frame shift

DNA sequence = TAC CAC (T)GG GTG ATT AGC T

mRNA sequence = AUG GUG ACC CAC UAA UCG A

amino acid sequence = Met / Val / thr / Gln / STOP

} Point mutation by insertion and translation sequence (18 nucleotides)

↳ Not only did the frame shift cause the val, and 3 amino acids to change; but also caused a nonsense mutation

↳ Proteins are packaged in certain ways depending on the amino acids & even the angles they are attached causing the protein no longer to function which can be fatal depending on what the protein does within the body ✓

this new sequence will undergo all the normal processes for protein synthesis but in the end the shape is incorrect

Mutations within DNA are permanent & can lead to health effects but all need medication, etc to help the person live a normal life

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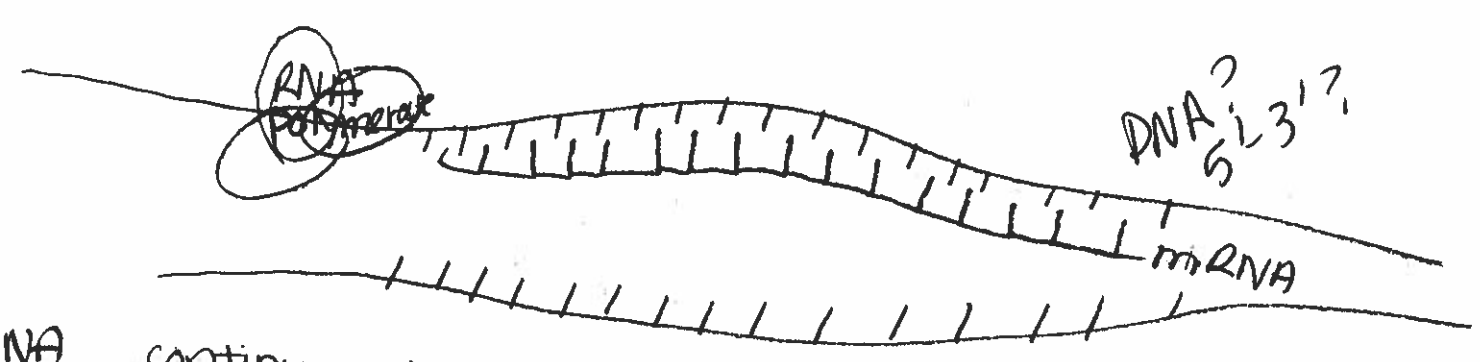
ID: A12

K 11/25 T 6/10 C 7/10 A 9/15 33/60

part C: The Big Question

In order to become a functional protein it must go through the processes of transcription and translation.

At the start of transcription RNA polymerase binds to the promotor of the DNA unzipped? coding strand vs non coding which is? template strand where the process of constructing mRNA will begin. The mRNA is what will be used to form the protein once it is translated.



The mRNA continues to elongate until it reaches a stop codon, which is? which causes the RNA polymerase and mRNA to dissassociate from the template strand. In eukaryotes the mRNA is then modified by the addition of a 5' cap to protect it from digestion and a poly A tail that protects the 3' end from degradation. ✓

The precursor mRNA is then made mature mRNA which is mostly non coding DNA. This is done by removing the introns (genes not expressed) how? splits? by cutting them out using spliceosomes and attaching the exons (expressed genes) to form the mature mRNA.

no! you are confused!

The mRNA is then refered to as tRNA (transport RNA) once it has entered the ribosomes for translation. the ribosomal subunits consist of the P-site, A-site, and E-site where protein chains are built.

The p-site forms polypeptide bonds, the A-site forms AA bonds on the chain, and the e-site is where the protein chain exits to the cytoplasm.

The tRNA is translated by its codons (3 letter nucleotides) which forms an amino acid.

Eg. AUG
MET

unclear how tRNA elongates the protein in the sites

10=31-40

During translation mutations can occur if a nucleotide is substituted, or deleted.

Esté?

The strand: AUG UGG GCA CAG ACC
MET TRP Ala Arg Thr

where all the peptide linkages?

which we'll say codes for hemaglobin can be altered if a mutation occurs.

If the letter A in the 3rd codon changed to a C the protein will not change (silent mutation)

$$\underset{\text{Ala}}{GCA} = \underset{\text{Ala}}{GCC}$$

you were asked for a mutation that makes a nonfunctional protein

However, if the letter U in the 2nd codon changed to a C the protein will change (missense mutation)

$$\underset{\text{Trp}}{UGG} \neq \underset{\text{Arg}}{CGG}$$

This would change the overall protein change and would end up coding for something other than hemaglobin.

Lastly, if a letter changed that ended up telling the protein to "stop" the protein would no longer be functional (nonsense mutation)

AUG UGG GCA CAA
Met Trp Ala STOP

unclear what then happens to the sequence to make a protein

unclear how these amino acids will change the protein

20/30 terms