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Block B

Mutations Activity

Part 1: Belgian Blue Myostatin

Pre-lab Questions:

1. Myostatin is found in the skeletal muscle of mammals
2. A growth factor is a molecule that controls parts of the cell cycle
3. "Knockout Mice" are mice that received myostatin
4. A mutation that will have no effect on an individual is called a silent mutation.
5. The most serious mutation is probably a frame-shift mutation because a frame-shift mutation will cause a change in all amino acids coded for in the mRNA causing a different protein to form.

Procedure:

1. Normal Myostatin:

DNA	TGT	GA**T**	**GAA**	**CAC**	**TCC**	**A**CA	GAA	TCT	CGA	TGC	TGT	CGC	TAC	CCC	CTC	ACG
mRNA	AC A	CUA	CUU	GU G	AG G	UGU	CUU	AGA	GCU	ACG	AGA	GC G	AUG	GG G	GA G	UGC
Amino Acid	Thr	Leu	Leu	Val	Arg	Cys	Leu	Arg	Ala	Thr	Arg	Ala	Met	Gly	Glu	Cys

2. Belgian Blue Myostatin:

DNA	TGT	GAC	AGA	ATC	TCG	ATG	CTG	TCG	CTA	CCC	CCT	CAC	GGT	GGA	TTT	TGA
mRNA	AC A	CUG	UCU	UAG	AGC	UA C	GAC	AGC	GAU	GG G	GG A	GU G	CCA	CCU	AAA	ACU
Amino Acid	Thr	Leu	Ser	Stop	Ser	Tyr	Asp	Ser	Asp	Gly	Gly	Val	Pro	Pro	Lys	Thr

3. (see bolded bases)

Analysis Questions:

1. The function of Myostatin is probably to limit muscle growth in mammals. This can be concluded from the fact that Belgian Blue and "knockout mice" have the mutated and therefore <u>inactive</u> myostatin and grow large muscles.
2. A deletion mutation occurred in the Belgian Blue myostatin.
3. A total of 11 base-pairs were deleted.
4. A nonsense mutation occurred because a stop codon was created in the base-pair sequence for Belgian Blue myostatin,
5. While Primary and Secondary structure are affected because a change in the amino acid sequence has occurred, the most notably change would be in Tertiary structure. Tertiary structure would be the most changed because a change in the amino acid will change how the amino acid R groups interact during the folding of the protein.

6. The mutation that occurred in Piedmontese cattle was a missense mutation because only one coded for amino acid changed due to the mutation.
7. Tertiary structure because a change in amino acids changes the R groups on the amino acids causing the interactions between R groups to be different, causing a different means of folding in tertiary structure, therefore changing the protein's function.

Part 2: Sickle Cell Anemia

Pre-lab Questions:

1. Hemoglobin is found in the red blood cells of eukaryote mammals.
2. Hemoglobin moves oxygen from the lungs to the body and carbon dioxide from the body to the lungs. (body here refers to all cells in the body)
3. Hemoglobin is made up of two alpha-globin subunits and two beta-globin subunits.
4. A normal red blood cell looks like a uncooked cookie dough that has a "smushed" center area while a sickle red blood cell looks like a sickle, resembling the letter "C".
5. Anemia is a condition where an individual has fewer than would be expected red blood cells in their body. Sickle Cell Anemia is an anemia because the sickle red blood cells will only live for 10-20 days while unaffected red blood cells will live for 120 days. This means that the sickle red blood cells will decrease in number over time if the rate of production of red blood cells is the same as would be in an unaffected individual.

Procedure:

1. Normal Hemoglobin:

DNA	CAC	GTG	GAC	TGA	GGA	C**T**C	CTC
mRna	GUG	CAC	CUG	ACU	CCU	GAG	GAG
Amino Acid	Val	His	Leu	Thr	Pro	Glu	Glu

2. Sickle Cell Hemoglobin:

DNA	CAC	GTG	GAC	TGA	GGA	C**A**C	CTC
mRna	GUG	CAC	CUG	ACU	CCU	GUG	GAG
Amino Acid	Val	His	Leu	Thr	Pro	Val	Glu

3. (See bolded bases)

Analysis Questions:

1. This is a substitution mutation.
2. 1 base-pair was changed.
3. This is a missense mutation because a base-pair was changed resulting in the changing of one coded for amino acid.
4. African-Americans, originated in Africa, a country with a widespread malaria problem. Since the sickle cell trait provides a resistance to malaria, Africans would develope this mutation to help combat malaria. This trait however runs the risk of having a child with sickle cell anemia if both parents have the trait. When Africans were moved to The United States, they kept their sickle cell traits and probability of sickle cell anemia that they had developed to resist malaria. Other races had no need to develop something like the sickle cell trait.
5. Sickle Cell Anemia is probably more common now because the affected individuals live longer, long enough to have children. If an individual with the Sickle Cell trait has children with a person with Sickle Cell anemia, there is a 50% chance of having an affected child, and a 100% chance that the children will at least have the trait. This is much higher than what would happen in the past when affected individuals did not live long enough to reproduce. With only individuals with the trait able to reproduce, there is only a 25% chance of having a child with Sickle Cell Anemia, much less than the 50% chance due to affected individuals living longer. (Note: In this explanation “affected individual” means a person with Sickle Cell Anemia)
6. Mutation Example:

	Original	**New**
DNA	**G**CC	**T**CC
mRNA	CGG	AGG
Amino Acid	Arg	Arg

This is an example of a silent mutation because the amino acid coded for will not change.