STUDY GUIDE - *Ch. 17.5 - Mutations of One or a Few Nucleotides can Affect Protein Structure and Function*

NAME: _____

- **PHYSICALLY PRINT OUT this PDF and HANDWRITE (with a black or blue pen) your answers directly on this PDF.** *Typed or digitally-written work is **not** accepted. Do **not** answer questions on separate paper.*
- **Importantly, study guides are NOT GROUP PROJECTS!!!** *You, and you alone, are to answer the questions as you **read** your assigned textbook. You are **not** to share answers with other students. You are **not** to copy any answers from any other source, including the internet.*
- **Get in the habit of writing LEGIBLY, neatly, and in a medium-sized font.** *AP essay readers and I will skip grading anything that cannot be easily read so start perfecting your handwriting, and don't write so large you can't add all the relevant details and key elaborations in the space provided.*
- **SCAN physical documents in color and with good resolution. Then, upload your final work as PDFs to Archie.** *Avoid uploading dark, shaded, washed-out, sideways, or upside-down scans of homework. Keep completed physical study guides organized in your biology binder to use as future study and review tools.*
- **READ FOR UNDERSTANDING and not merely to complete an assignment.** ***First**, read a section quickly to get an overview of the topic covered. Then, read it a **second** time slowly, paraphrasing each paragraph **out loud** and analyzing every figure. Finally, read it a **third** time as you answer the study guide questions if assigned and start building your memory. Try to write answers out in your own words, when possible, and try to purposefully and accurately use all new terminology introduced.*

Mutations are the ultimate and original source of new alleles of genes and even new genes!!!

We studied **gross (large-scale) mutations** that can involve one or many genes and large pieces of chromosomes - *deletions, insertions, inversions, translocations* - already, which often happen because of **errors like non-disjunction errors during anaphase in mitosis or anaphase I or II in meiosis** or because of **errors during crossing-over** during prophase I of meiosis. Review these by revisiting Ch.15.4!

Now you must understand more **microscopic mutations** too - *deletions, insertions, substitutions (and potential frame shifts that occur when ribosomal reading frames are altered)* -, many of which can also form because of **errors by DNA Polymerase during S phase DNA replication**, ***damage to DNA template strands by mutagens or radiation***, as well as smaller errors during DNA recombination when **crossing-over occurs** in meiosis I.

1. What is a **mutation** in terms of molecular genetics?

2. What are **point mutations**?

3. **IMPORTANTLY:** Not all mutations that may occur in a cell in one organism enter the **gene pool** of the next generation *(the collection of alleles for all the genes in all organisms in a population)* even if that mutated cell can produce daughter cells that do inherited the parent cell's DNA mutation. (Think of a transformed skin cell hat became cancerous after key tumor suppressor and proto-onco genes became mutated following DNA damage caused by U.V. radiation, for example. These cancerous skin cells may now divide and pass down the DNA mutations to their daughter cells (forming a tumor), the descendant cells all being cancerous too and dividing uncontrollably themselves. Yet, these cancerous skin cell's DNA mutations are **not** passed down to the zygote if this individual mates since skin cells don't engage in meiosis to make the organism's reproductive cells or gametes). So, **some mutations occur in an organism's cells, but still cannot be passed down to the offspring of that individual organism.**

   When is a **DNA mutation passed down to future generations**? Where does the mutation have to appear?

4. What changes in the DNA when the **point mutation** known as a **nucleotide-pair substitution** or **base-pair substitution** occurs?

5. **Two types of genes exist in cells:** RNA genes that code for RNA molecules and Protein genes that code for a polypeptide of a protein (and, thus, for proteins).

   Mutations that occur in **controlling elements** **of genes do not alter the nucleotide sequence of RNA molecules or the primary structure of proteins (and therefore do not alter the shape and function of a resulting RNA molecule or protein)** like mutations that occur in the transcription unit of a gene may do. However, mutations in controlling elements of genes (promoters and enhancers in eukaryotes or promoters or promoters and operators in prokaryotic single genes or operons, respectively, **can influence if a gene can be expressed at all or not, or how much and when a gene is expressed. By influencing the concentration and timing of protein construction, the phenotype of a cell may be altered, even if the same or "correct" version of a protein or RNA is made** as was happening before this mutation in the controlling element of the gene occurred.

   Of course, changes in the **transcription unit** (open reading frame) of a gene in prokaryotes and in the **exons** (transcription unit coding sequences) of a gene in eukaryotes, **will alter the nucleotide sequence in the RNA made and may alter the primary structure of a polypeptide making up a protein and thus potentially alter the RNA or protein's 3-D shape and thus its function.** I say may alter the proteins primary structure since some mutations in the transcription unit (and exons in eukaryotes) may still result in the cell putting down the same amino acid in the polypeptide as before due to the redundancy in the genetic code (multiple codons existing for a particular amino acid).

   Remember that mutations in eukaryotic transcription unit's **introns** will be copied into the pre-mRNA but will be **removed from the mature mRNA** transcript before the ribosome uses the information to construct a protein so the intronic noncoding sequences would **not affect protein shape and function**, though research shows that **some introns may play a role in influencing gene expression.** Changes in these introns may, thus, also alter when and if that gene can be expressed appropriately.

   It is **imperative** that you thoroughly understand and have FULLY MEMORIZED **Figure 17.27** and the text related to this topic so study this image carefully as your read on.

   a. What **type of base-pair substitution is a silent mutation**?

   b. What **type of base-pair substitution is a missense mutation**?

   c. What type of base-pair substitution is a **nonsense mutation**?

6. a. What type of point mutation is an **insertion**?

   b. What type of point mutation is a **deletion**?

7. Though some mutations are beneficial or neutral to a cell or to an organism's phenotype (up to ~20% of mutations), most often mutations will be harmful (~80% of mutations). **Insertions and deletions often (though not always) have more disastrous effects on the phenotype of a cell or organism compared to single-nucleotide substitutions.** This is because they could potentially alter the reading frame of the resulting mRNA transcribed from a gene that experienced these types of mutations *(depending on which and how many nucleotides are involved in the insertion or deletion mutation of course).*

   a. Mutations that alter reading frames of genes, and so certain insertion or deletion mutations (but not all automatically), are also called **frameshift mutations**. What does it meant that this mutation **altered the "reading frame"**?

   b. What change in the DNA would most likely result in a **frameshift mutations**?

   c. One of these insertion or deletion mutations **is** a frameshift mutation and two of these insertion or deletion mutations are **not** a frameshift mutation. Transcribe the DNA template into mRNA and then translate the mRNA into a polypeptide chain using the **Codon Table in Figure 17.6**. Then, determine if the mutation was an insertion but not a frameshift mutation, a deletion but not a frameshift mutation, an insertion and a frameshift mutation, a deletion and a frameshift mutation. *Don't forget to label the 3', 5', N - , C - ends of your polynucleotides and your polypeptides.*

	Wild Type Allele of Gene	**DNA Mutation #1**
DNA Template Strands	3'-GCTTATGCACAATGGTTT-5'	3'-GCTGGGTATGCACAATGGTTT-5'
mRNA		
Amino acid sequence		

Is this a DNA insertion or deletion mutation? _____ Is this a DNA frameshift mutation? _____

	Wild Type Allele of Gene	**DNA Mutation #2**
DNA Template Strands	3'-GCTTATGCACAATGGTTT-5'	3'-GTTATGCACAATGGTTT-5'
mRNA		
Amino acid sequence		

Is this a DNA insertion or deletion mutation? _____ Is this a DNA frameshift mutation? _____

	Wild Type Allele of Gene	**DNA Mutation #3**
DNA Template Strands	3'-GCTTATGCACAATGGTTT-5'	3'-GCTTCGAATGCACAATGGTTT-5'
mRNA		
Amino acid sequence		

Is this a DNA insertion or deletion mutation? _____ Is this a DNA frameshift mutation? _____

8. Though **a gene is typically thousands of nucleotides long**, suppose a gene whose template strand contains the sequence 3′-TACTTGTCCGATATC-5′ is mutated to 3′-TACTTGTCC**A**ATATC-5′. **This now comprises a new allele of the gene in a population if it is passed down in the gamete to the next generation**.

   a. For both normal and mutant alleles, draw the double-stranded DNA, the resulting mRNA, and the amino acid sequence each encodes side-by-side below. *As always, be sure to label the 3′, 5′, N - , and C - ends of your polynucleotides and your polypeptide.*

	Wild Type Allele of Gene	**Mutant Allele of Gene**
DNA Template Strands *(the strand that **IS** transcribed)*	3′-TACTTGTCCGTCATCACT-5′	3′-TACTTGTCC**A**TCATCACT-5′
DNA Coding Strands *(the strand that is **NOT** transcribed)*		
mRNA		
Amino acid sequence		

   b. What is the **effect of this mutation on the amino acid sequence**?

   c. What **type of a mutation** was this?

9. a. What are **chemical and physical mutagens**?

   b. What is are two examples of a **physical mutagens**?

      1. _____ 2. _____

   c. Describe the actions of three different types of **chemical mutagens**.

      1.

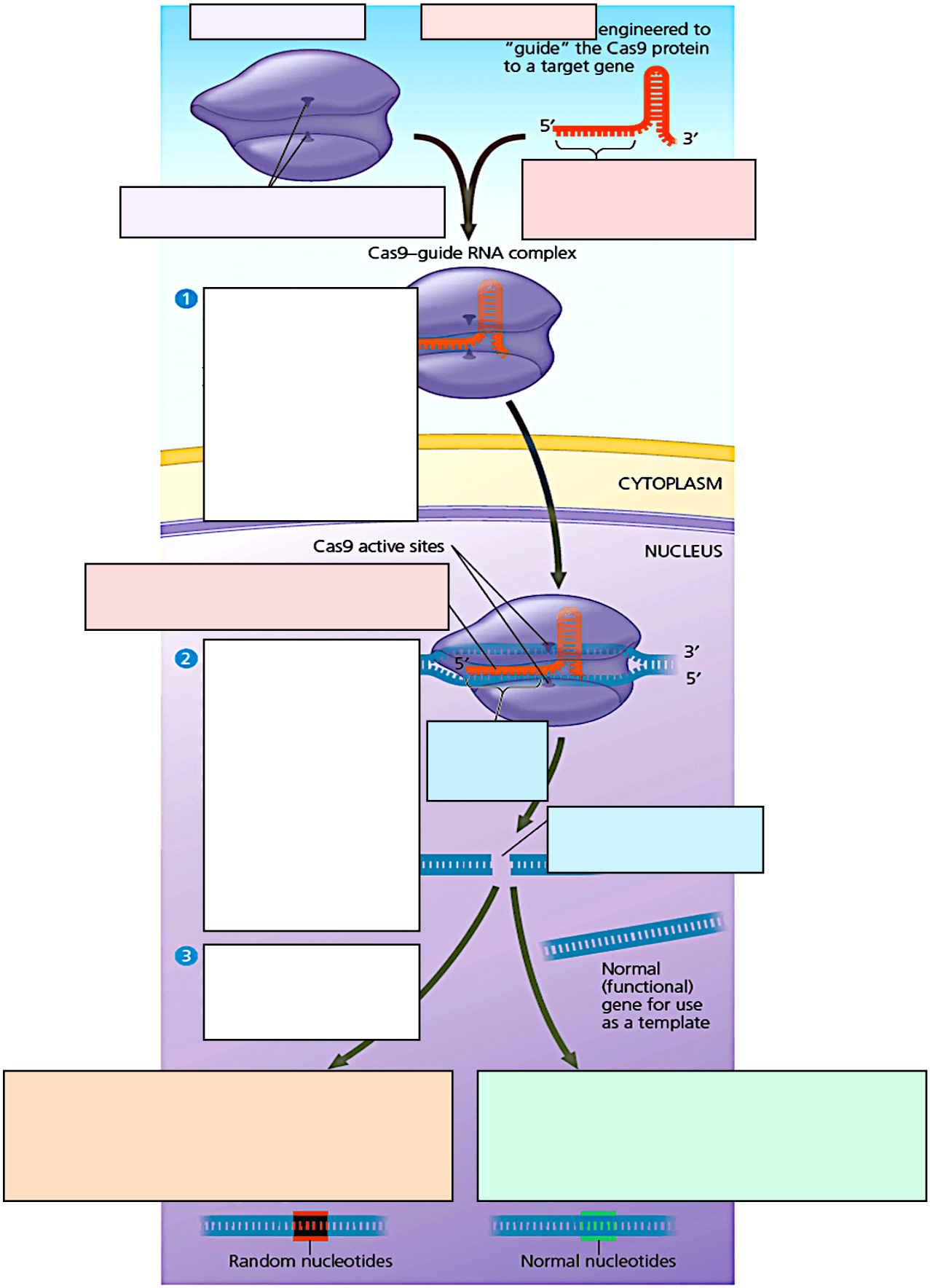
      2.

      3.

10. a. What does **gene editing** involve?

    b. What is **genetic engineering**?

11. **CRISPR-Cas9** is a defense system against viruses discovered in bacteria, which scientists are now manipulating to use in any cell in order to introduce changes into DNA sequences (either **random mutations** or **specified nucleotide changes**). Read through this section of your text to understand CRISPS-Cas9's uses better, then label the image to outline the process by which scientists use this system to purposely alter genes in chromosomes of cells.



12. Knowing all we know about genetics today, what is the best definition we have currently for **what is a gene**?

13. Proceed to the TEST YOUR UNDERSTANDING section at the end of the chapter. **Study your chapter sections and all Ch.17 study guides first!** Then, do your best to try to answer these from memory first in order to test how well you grasped the material before. If you are unsure, return to the relevant section of your chapter and restudy any pertinent material to refresh your memory. *(Check some of your answers by going to the Ch.17 Test Your Understanding answers in Appendix A)*

1. _____ 2. _____ 3. _____ 4. _____ 5. _____ 6. _____ 7. _____

8.

9. Fill in the Missing Parts of the Table.

Type of RNA	Function
Messenger RNA (mRNA)	
Transfer RNA (tRNA)	
	Found in a ribosome. Plays both a structural role and as a ribozyme also plays a catalytic role (catalyzing peptide bond formation)
Primary Transcript	
Small RNAs in Spliceosomes	

10.

13.