Chapter Review

1. Describe the major contributions of the following scientists regarding the discovery of the structure of DNA.

Hershey and Chase Working with bacteriophages, these scientists used transformation experiments to confirm that DNA, rather than proteins, is the genetic material. They discovered that the DNA of the virus infects the bacterial cell and changes its genetic code, destroying the cell after having turned it briefly into a virus factory.

Erwin Chargaff He noted that the DNA of different species, as well as from different sources within the same organism, showed certain chemical regularities. Specifically, he found that the amount of guanine equaled the amount of cytosine (G=C), and that the amount of adenine equaled that of thymine (A =T). Correspondingly, the total amount of purines (A+G) always equaled total pyrimidines (A+G =T+C). This is called "Chargaff's rule" and was an important step leading to the discovery of the structure of DNA.

Rosalind Franklin This chemist discovered the crucial evidence regarding the structure of DNA through experiments using x-ray crystallography. Her crystallographs enabled scientists to visualize the helical structure, a spiral shape like a spring, of the DNA molecule.

Watson and Crick They brought together all relevant evidence to create the first double helix model of the DNA molecule. They suggested that nucleotide bases are on the interior of the two strands, a sugarphosphate backbone is on the outside, and that base pairs have a uniform width along the double helix.

2. Define bacterial transformation and discuss how studies of this phenomenon influenced DNA research.

Bacterial transformation is a mechanism that results in the transfer of genetic information between different bacterial cells. In transformation, DNA from a bacterium of one genotype is taken in through the cell surface of another bacterium and then the imported DNA is incorporated into the chromosome of the recipient cell. Transformation also occurs in eukaryotes, and experiments on multicellular eukaryotes provided experimental evidence for DNA as the genetic material.

3. In living organisms, the amount of adenine is equal to the amount of thymine, and the amount of cytosine is equal to that of guanine. A researcher measured the amount of adenine in a cell, and found it to be 15% of the DNA. Calculate the percent amount of the remaining nucleotides.

Thymine = 15%, guanine = 35%, cytosine = 35%.

4. Explain why the ratio of A+T:C+G is always the same within a single species, yet differs across more than one species.

The ratios are the same within a single species, because the particular ratios and arrangements of nucleotides allow sexual reproduction between individuals of a certain species. During meiosis, chromosomes from the father must be capable of pairing with like chromosomes from the mother, and this requires the same pattern and ratio of nucleotides. Most members of the same species are identical in the vast majority of their genomes. Across different species, however, the ratios differ, reflecting the multitude of genetic differences that distinguishes one species from another.

5. Explain how the double helical structure of DNA allows:

a. Storage of genetic information.

Because DNA contains millions of nucleotides, the base sequences of DNA molecules can encode and

store enormous amounts of information. Variations in DNA sequences account for genetic differences among species and individuals.

b. Precise replication during the cell division cycle.

The specific pairing of A with T and of G with C provides an exact copying mechanism for the genetic material during the cell division process.

c. Susceptibility to mutations.

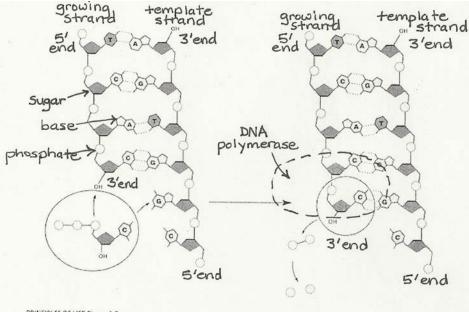
The structure of DNA, when it undergoes simple changes in the linear sequence of base pairs, is said to have undergone a mutation. With two strands, a mutation on one strand can be overcome if the other strand is not altered.

d. Expression of the coded information as phenotypes.

To make a phenotype, the nucleotide sequence of DNA is copied into RNA, with the linear sequence of nucleotides in the RNA is later translated to coordinate the synthesis of a linear sequence of amino acids. These folded forms of proteins determine many of the phenotypes of an organism. Precision unfolding of the double strands is required before the "sense" strand is used to direct RNA synthesis.

6. The diagram below shows a strand of DNA being replicated.

Label the following: a phosphate, sugar, nitrogenous base, DNA polymerase, growing strand, and template strand. For each strand, label the 5' and 3' ends.



PRINCIPLES OF LIFE. Figure 9.7

7. In the diagram above, the strand on the left shows the addition of a cytosine with three phosphates attached to it. Two of the phosphates will ultimately become detached. What result is achieved by the departure of the two phosphate groups?

Bonds linking the phosphate groups are broken, releasing energy to drive the reaction.

8. Briefly describe the function of each of these three enzymes.

Primase: This is an enzyme that catalyzes the synthesis of a primer for DNA replication.

DNA Polymerase: Any of a group of enzymes that coordinates the formation of DNA strands from a DNA template.

Ligase: A specific enzyme that facilitates the fusion of DNA strands by catalyzing the formation of a phosphodiester bond, playing an important role in repairing missing or lagging fragments of DNA.

9. Discuss continuous and discontinuous replication, using the terms, "leading strand," and "lagging strand."

One newly synthesized strand, the leading strand, is oriented so that it can be replicated continuously beginning at its 3' end as the fork opens up. The other new strand, the lagging strand, must be synthesized differently because it grows in the opposite direction, away from the replication fork. The synthesis of the lagging strand consists of discontinuous stretches of new DNA called Okazaki fragments.

10. Compare the point of origin of DNA replication between eukaryotes and prokaryotes, and explain how this difference serves an important function.

As prokaryotes have circular chromosomes, there is a single origin of replication per circular chromosome. Eukaryotes, however, have linear chromosome, so there are multiple origins of replication on each linear chromosome, and they initiate the replication process at different times. This helps speed the duplication time, which is important because of eukaryotes' much larger amount of genetic material compared to prokaryotes.

11. Explain how adjacent Okazaki fragments become linked together to form a continuous strand of DNA.

While a single primer is needed to initiate the synthesis of the leading strands, each Okazaki fragment requires its own primer to be synthesized by the primase. DNA polymerase synthesizes an Okazaki fragment by adding nucleotides to one primer until it reaches the primer of the previous fragment. At this point, a different DNA polymerase removes the old primer and replaces it with DNA. This leaves a tiny gap, the final phosphodiester linkage between the adjacent Okazaki fragments. DNA ligase catalyzes the formation of that bond, linking the fragments and making the lagging strand whole.

12. Explain how the ends of a chromosome are shortened each time a chromosome replicates and describe how telomeres help prevent the loss of genetic material.

Many eukaryotes have strings of repetitive sequences called telomeres at the ends of their chromosomes that bind special proteins to the ends of these chromosomes. As a certain amount of telomeric DNA is lost in each round of replication and cell division, the telomeres on the ends of the strands become gradually shortened, but this help protects the genetic material further toward the middle of the chromosome. The chromosome ends can eventually become short enough to lose these protective fragments, causing the chromosomes to ultimately lose their integrity. This is a possible aging or senescence characteristic of eukaryotic life.

13. Define the function of telomerase and describe what types of cells are particularly dependent on its continual function.

Telomerase is an enzyme that catalyzes the addition of telomeric sequences lost from chromosomes during DNA replication. Continuously dividing cells, like bone marrow stem cells and gamete-producing cells, can only maintain their protective telomeric DNA if telomerase is functioning properly.

14. Describe the mechanism by which PCR proceeds, and discuss PCR's use.

PCR, or polymerase chain reaction, allows researchers to make multiple copies of short DNA sequences in a test tube in a process called DNA amplification. This technique is used to analyze DNA, genes, and genomes. PCR proceeds by repetition of a sequence of steps over and over. The specificity of the DNA sequences is a

key to the power of PCR to amplify a small part of a larger DNA molecule. This process is useful for identification of individuals and detection of disease.

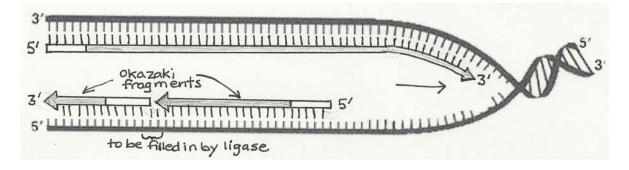
15. Assume you need to amplify (copy) a single gene from a eukaryotic organism with 8 chromosomes. Describe the "ingredients" you would need and state the function of each ingredient.

- A sample of double-stranded DNA (to act as the template)
- Two short, artificially synthesized primers that are complementary to the ends of the sequence to be amplified (these are made under laboratory conditions based on the DNA sample)
- The four dNTPs (dATP, dTTP, dCTP, and dGTP), i.e., the four nucleotide components of DNA
- A DNA polymerase that can tolerate high temperatures without becoming denatured. This enzyme will catalyze the formation of DNA strands from the template.
- Salts and a buffer to maintain a new-neutral pH, i.e., the optimal condition for stability of the enzymes and the nucleotides.

16. The diagram below shows two strands of DNA being replicated.

a. Draw in the DNA on the continuous side being formed by DNA polymerase.

b. Draw in two Okazaki fragments on the discontinuous side, one formed, and the second still being formed by DNA polymerase. Label the spot to be filled in by ligase.



17. When a person develops skin cancer as an adult, is this caused by a somatic mutation or germ line mutation? Explain your answer.

Skin cancer is caused by a somatic mutation, one that occurs in a somatic cell and cannot be inherited through sexual reproduction. A germ line mutation could, nevertheless, make getting skin cancer more likely, if an individual were to inherit a mutation in the genes whose expression suppress cancer.

18. Explain the difference between silent and loss-of-function mutations, and explain which type is more commonly seen.

The most common type of mutations are "silent" mutations. They are so-called because these mutations do not affect gene function, either because it is not expressed or because the nucleotide change does not have any effect on the encoded protein. Loss-of-function mutations result in either loss of expression of a gene or in the production of an RNA that leads to a nonfunctional "mutant" protein.

19. Describe and discuss the differences between point mutations and chromosomal mutations.

A point mutation is caused by the gain, loss, or substitution of a single nucleotide, while a chromosomal mutation is the loss of or changes in position or direction of a DNA segment on a chromosome. A point mutation within a gene can cause a new allele that may or may not result in a new phenotype. Chromosomal mutations are major changes in chromosomal structure, which can have severe consequences.

20. Identify 5 different mutagens that are in your environment and indicate how you might avoid each.

a. Benzopyrene in cigarette smoke – avoid by not smoking!

b. Ultraviolet radiation -avoid excess exposure to sunlight, use sunscreen, wear protective clothing

- c. Nitrates in processed meats avoid eating them, or consume in conjunction with ascorbate
- d. DDT (agricultural insecticide) support ban on its use in all nations; avoid produce where DDT is used
- e. Asbestos avoid building materials made of asbestos, hire OHSU certified workers to remove it

21. Describe the type of mutations that provide the raw material for natural selection. Explain your answer.

Mutation generates the genetic diversity that makes natural selection possible. A mutation in a germline cell may not have an immediate selective advantage, but it may cause a phenotypic change in the offspring which provide a better adaptation for later environmental changes.

22. Discuss how is PCR used to examine the DNA of Neanderthals.

DNA from the 50,000-year-old bones of skeletons of Neanderthals has been extracted and amplified by scientists, using the PCR process, allowing them to study the entire Neanderthal DNA sequence.

23. If you search the Internet for images of Neanderthals, you will find many older images that depict Neanderthals as dumb, ape-like, and inferior to modern humans. Yet figure 9.20 in your text (shown to the right) depicts a Neanderthal looking very much human-like. Explain why.

Scientists who have been studying the genome from cells taken from the bones of Neanderthal skeletons have discovered that their DNA sequence is over 99 percent identical to that of modern humans. Specific gene comparisons include those for hair and skin, which suggest that Neanderthals may have had fair skin and reddish hair. The Neanderthal woman depicted to the right looks remarkably human-like. However, given the tremendous amount of similarity in the genome of human and Neanderthals, these similarities should be expected rather than surprising.



Science Practices & Inquiry

24. In the 30 April 2010 issue of **Science**, Roach, J. C., *et al.*, reported that the mutation rate for **humans** is approximately 1.1×10^{-8} mutations per base pair in the haploid genome. Humans have a diploid genome of 6 x 10^{9} base pairs.

a. Calculate the number of mutations in each new child. Show your work.

 $(1.1 \times 10^{-8} \text{ mutations/haploid base pair}) \times 2 \text{ haploid base pairs/diploid base pair}$

= 2.2×10^{-8} mutations/diploid base pairs

 $(2.2 \times 10^{-8} \text{ mutations/diploid base pair}) \times (6 \times 10^{9} \text{ diploid base pairs}) = 13.2 \times 10^{1} = 132 \text{ mutations/child}$

b. These are spontaneous mutations. Explain why the majority of these mutations have no effect on a new organism.

Spontaneous mutations are permanent changes in the genetic material that occur without any outside influence. Since the DNA replication process contains a "proof-reading" function, the majority of these errors are corrected. Even those that evade correction may not show up in the phenotype of the new organism, and those that do may not result in any harmful effects.