20.4 MUTATIONS AND GENETIC VARIATION

Case Study: Gene Mutations and Cancer

(Pages 689–690)

1. Cancer is the result of altered genetic information. Viruses inject foreign genetic material into host cells. Some cancers are closely associated with specific viral diseases (e.g., genital warts and cervical cancer). Therefore, some viruses probably trigger cancer.
2. Ultraviolet radiation produced as part of solar radiation will damage skin cells, particularly the chromosomes. The genetically damaged cells are prone to becoming cancerous. When the skin is protected from UV radiation, there is a significant decrease in skin cancer cases.
3. Tar products: building/repair industry; cigarette smoke: smoking; UV radiation; sunlight
4. An oncogene causes the specific cancer. A proto-oncogene is an oncogene in an inactive form, which must be initiated to become an active oncogene.
5. Oncogenes may be activated by carcinogens in the environment, which may mutate the genes. It probably takes a number of "hits" to the cell's genetic makeup to finally activate an oncogene.
6. The *ras* gene is a common proto-oncogene that normally controls cell division in tissues. If activated as an oncogene, it can no longer carry out this function, so cells divide uncontrollably. The *ras* gene is found in 50 % of colon cancers.

Lab Exercise 20.B: Looking for SINEs

(Pages 691–692)

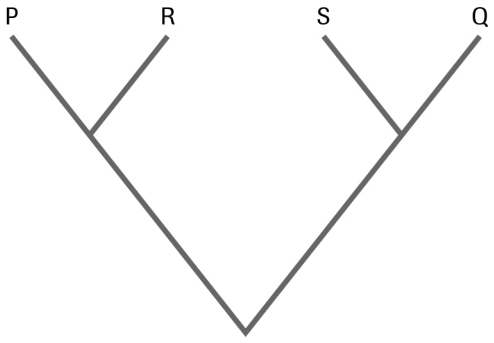
Analysis

Part I: Looking for a SINE

(a) There is a single nucleotide difference. This could have resulted from a point mutation happening any time after an original insertion event.

(b) These are likely to be neutral as the SINE insertion is in a noncoding section of DNA.

(c) The following diagram illustrates the relationship between these species suggested by the data. The SINE is evidence of a common shared ancestor of species Q and S. Species P and R are very similar and therefore likely diverged more recently.



Part II: Evolution Displayed by SINEs and LINEs

(d) Whales are more closely related to hippos, as indicated by the additional two shared SINEs, C and H. If two species share even one identical SINE insertion, it is highly probable that a shared common ancestor had the insertion. It is extremely unlikely that they experienced an identical mutation event of this type independently.

(e) Insertion B must have happened first as four species have inherited this insertion, whereas only two of them have inherited insertion A. This suggests that the insertion event for SINE A occurred after a lineage containing B had split.

Synthesis

(f) Pigs and camels are equally closely related to camels. They both share the same most recent common ancestor (at the very base of the cladogram diagram).

(g) All whales must possess SINEs B, G, C, H, and I. As indicated in the cladogram, these five SINEs all evolved in direct ancestors of the whales. The SINEs D, E, and A evolved in groups that had diverged from the ancestral whale lineage, whereas SINE F evolved after the whale lineage had separated from hippo lineage. Students should also realize that *no* whale could possess any of the other SINEs here.

(h) The researcher could explore the genomes of all three species. If they found a SINE shared by only two of the three species, they could conclude that they shared a more common ancestor. It is possible that orcas are equally closely related to white-sided dolphins and pilot whales. Such a situation would be confirmed by finding a SINE shared by the pilot whales and dolphins *not* found in the orca.

Section 20.4 Questions

(Page 694)

1. mutation: a change in the DNA sequence that is inherited
   frameshift mutation: a mutation that causes the reading frame of codons to change, usually resulting in different amino acids being incorporated into the polypeptide
   point mutation: a mutation at a specific base pair in the genome
   nonsense mutation: a mutation that converts a codon from an amino acid into a termination codon
   missense mutation: a mutation that results in the single substitution of one amino acid in the resulting polypeptide
2. Nitrogen-base additions can be more harmful than nitrogen-base substitutions because a nitrogen-base substitution can result in the expression of a different amino acid codon. Depending on where the amino acid falls within the protein, it may play a minor role in altering protein performance. A nitrogen-base addition changes the reading frame, resulting in a completely new sequence of amino acids being expressed, which can render the protein inactive. There are some exceptions to the nitrogen-base substitution scenario. If a nitrogen-base substitution results in a nonsense mutation, the results are similar to a nitrogen-base addition scenario.
3. A nonsense mutation is more harmful than a missense mutation. A nonsense mutation results in the termination of translation; therefore, the protein that is to be translated is not fully synthesized, rendering it inactive. A missense mutation results in one amino acid change. The result is a protein that may or may not function to full capacity, depending on where the amino acid substitution has fallen. A missense mutation can be just as harmful as a nonsense mutation, but there is the slight chance that it will not.
4. Three factors that produce gene mutations are UV radiation, X-rays, and chemicals such as pesticides.
5. AUG–UUU–UUG–CCU–UAU–CAU–CGU (native strand)
   met–phe–leu–pro–tyr–his–arg
   (a) New protein: met–phe–leu–pro–tyr–his–arg
   The mutation has no effect since UAU and UAC both code for tyrosine.
   (b) New protein: met–phe–leu–pro–STOP
   The mutation is a nonsense mutation. UAA is a stop codon so the protein is not fully translated and is therefore nonfunctioning.
   (c) New protein: met–phe–leu–ala–leu–leu–leu
   The addition causes a frameshift mutation, resulting in different amino acids.

(d) New protein: met–phe–leu–pro–ile–ile
The deletion causes a frameshift mutation, resulting in different amino acids.
(e) New protein: cys–tyr–tyr–phe–val–phe–val
The inversion results in a different protein being synthesized.

6. (a) Arginine can change to leucine with the following substitutions: CGU to CUU, CGC to CUC, CGA to CUA, and CGG to CUG.
(b) Cysteine to glutamic acid cannot be changed with one base-pair substitution.
(c) Serine can be changed to threonine with the following substitutions: AGU to ACU and AGC to ACC.
(d) Isoleucine can be changed to serine with the following substitutions: AUU to AGU and AUC to AGC.
7. A food dye that has been identified as a chemical mutagen poses greater dangers for a developing fetus than for an adult. A fetus is undergoing rapid developmental growth within the uterus. The rate of mitosis for all cells is much faster than that within an adult. The effects of the mutagen can vary depending on which stage of development the fetus is in. If the mutagen affects nondifferentiated cells (cells that will eventually become specialized cells, such as liver, heart, kidney), it may impart a serious mutation that will lead to abnormal development. An adult also undergoes mitosis but does so to replace existing cells. If a mutation takes place in an adult, chances are it will be limited to the one cell and its daughter cells in the future; therefore, it will be localized to one area. In a fetus, a mutation will affect all cells since undifferentiated cells can become many different types of cells.
8. Students' answers will vary. Some suggestions include wearing sunscreen, not using pesticides on lawns, washing all food thoroughly before ingestion, and avoiding unnecessary X-rays.
9. Mutations result in changes in the type of proteins that are made by an organism. This could result in different structures of functions in existing proteins of the body. If the mutation is of benefit to the organism, it will be better adapted to its environment and will survive to pass on this mutation to its offspring. Natural selection influences which mutations remain in a population.
10. Students' answers will vary but should discuss the points noted here. The underlying theme of endosymbiotic theory was formulated in 1966 by Lynn Margulis, when she was a young faculty member at Boston University. Her 1970 book, *Origin of Eukaryotic Cells*, discusses her early work pertaining to this organelle genesis theory in detail. Currently, her endosymbiotic theory is recognized as the key method by which organelles could have arisen and is widely accepted by the mainstream scientific community. The endosymbiotic theory of organogenesis was actually proven in the 1980s, when the genetic material of mitochondria, centrioles, and chloroplasts was found to be different from that of nuclear DNA.

    The endosymbiotic theory concerns the origins of mitochondria and plastids (plastids with chlorophyll a and b are called chloroplasts; some other plastids are called cyanelles and rhodoplasts), which are organelles of eukaryotic cells. According to this theory, these originated as prokaryotic organisms, which came to live inside prokaryotic cells as endosymbionts. In other words, the endosymbiotic theory suggests that eukaryotic cells first appeared when a prokaryotic cell was absorbed into another cell without being digested. The theory also postulates that the mitochondria evolved from facilitated anaerobic bacteria (probably proteobacteria, related to the rickettsias) and that the chloroplast evolved from endosymbiotic cyanobacteria (autotrophic prokaryotes). The evidence for this theory is compelling as a whole, and it is now generally accepted.
11. Glutamic acid is a polar molecule, whereas valine is not. This may result in changes in the structure of the hemoglobin molecule.
    If other polar molecules were substituted, then the hemoglobin would not be altered. Glutamic acid and aspartic acid have a similar chemical structure.

