Learning Objectives:

LO 2.38 The student is able to analyze data to support the claim that responses to information and communication of information affect natural selection. [See **SP 5.1**]

LO 2.39 The student is able to justify scientific claims, using evidence, to describe how timing and coordination of behavioral events in organisms are regulated by several mechanisms. [See **SP 6.1**]

LO 2.40 The student is able to connect concepts in and across domain(s) to predict how environmental factors affect responses to information and change behavior. [See **SP 7.2**]

Note: Learning objectives 2.41, 2.42, and 2.43 are located after learning objectives 2.5, 2.21, and 2.30, respectively.

Big Idea 3: Living systems store, retrieve, transmit and respond to information essential to life processes.

Genetic information provides for continuity of life and, in most cases, this information is passed from parent to offspring via DNA. The double-stranded structure of DNA provides a simple and elegant solution for the transmission of heritable information to the next generation; by using each strand as a template, existing information can be preserved and duplicated with high fidelity within the replication process. However, the process of replication is imperfect, and errors occur through chemical instability and environmental impacts. Random changes in DNA nucleotide sequences lead to heritable mutations if they are not repaired. To protect against changes in the original sequence, cells have multiple mechanisms to correct errors. Despite the action of repair enzymes, some mutations are not corrected and are passed to subsequent generations. Changes in a nucleotide sequence, if present in a protein-coding region, can change the amino acid sequence of the polypeptide. In other cases, mutations can alter levels of gene expression or simply be silent. In order for information in DNA to direct cellular processes, information must be transcribed (DNA → RNA) and, in many cases, translated (RNA → protein). The products of transcription and translation play an important role in determining metabolism, i.e., cellular activities and phenotypes. Biotechnology makes it possible to directly engineer heritable changes in cells to yield novel protein products.

In eukaryotic organisms, heritable information is packaged into chromosomes that are passed to daughter cells. Alternating with interphase in the cell cycle, mitosis followed by cytokinesis provides

a mechanism in which each daughter cell receives an identical and a complete complement of chromosomes. Mitosis ensures fidelity in the transmission of heritable information, and production of identical progeny allows organisms to grow, replace cells, and reproduce asexually.

Sexual reproduction, however, involves the recombination of heritable information from both parents through fusion of gametes during fertilization. Meiosis followed by fertilization provides a spectrum of possible phenotypes in offspring and on which natural selection operates.

Mendel was able to describe a model of inheritance of traits, and his work represents an application of mathematical reasoning to a biological problem. However, most traits result from interactions of many genes and do not follow Mendelian patterns of inheritance. Understanding the genetic basis of specific phenotypes and their transmission in humans can raise social and ethical issues.

The expression of genetic material controls cell products, and these products determine the metabolism and nature of the cell. Gene expression is regulated by both environmental signals and developmental cascades or stages. Cell signaling mechanisms can also modulate and control gene expression. Thus, structure and function in biology involve two interacting aspects: the presence of necessary genetic information and the correct and timely expression of this information.

Genetic information is a repository of instructions necessary for the survival, growth and reproduction of the organism. Changes in information can often be observed in the organism due to changes in phenotypes. At the molecular level, these changes may result from mutations in the genetic material whereupon effects can often be seen when the information is processed to yield a polypeptide; the changes may be positive, negative or neutral to the organism. At the cellular level, errors in the transfer of genetic information through mitosis and meiosis can result in adverse changes to cellular composition. Additionally, environmental factors can influence gene expression.

Genetic variation is almost always advantageous for the long-term survival and evolution of a species. In sexually reproducing organisms, meiosis produces haploid gametes, and random fertilization produces diploid zygotes. In asexually reproducing organisms, variation can be introduced through mistakes in DNA replication or repair and through recombination; additionally, bacteria can transmit and/or exchange genetic information horizontally (between individuals in the same generation). Viruses have a unique mechanism of replication that is dependent on the host metabolic machinery. Viruses can introduce variation in the host genetic material through lysogenesis or latent infection.

To function in a biological system, cells communicate with other cells and respond to the external environment. Cell signaling pathways are determined by interacting signal and receptor molecules, and signaling cascades direct complex behaviors that affect physiological responses in the organism by altering gene expression or protein activity. Nonheritable information transmission influences behavior within and between cells, organisms and populations; these behaviors are directed by underlying genetic information, and responses to information are vital to natural selection and evolution. Animals have evolved sensory organs that detect and process external information. Nervous systems interface with these sensory and internal body systems, coordinating response and behavior; and this coordination occurs through the transmission and processing of signal information. Behavior in the individual serves to increase its fitness in the population while contributing to the overall survival of the population.

Enduring understanding 3.A: Heritable information provides for continuity of life.

The organizational basis of all living systems is heritable information. The proper storage and transfer of this information are critical for life to continue at the cell, organism and species levels. Reproduction occurs at the cellular and organismal levels. In order for daughter cells to continue subsequent generational cycles of reproduction or replication, each progeny needs to receive heritable genetic instructions from the parental source. This information is stored and passed to the subsequent generation via DNA. Viruses, as exceptional entities, can contain either DNA or RNA as heritable genetic information. The chemical structures of both DNA and RNA provide mechanisms that ensure information is preserved and passed to subsequent generations. There are important chemical and structural differences between DNA and RNA that result in different stabilities and modes of replication. In order for information stored in DNA to direct cellular processes, the information needs to be transcribed (DNA → RNA) and in many cases, translated (RNA → protein). The products of these processes determine metabolism and cellular activities and, thus, the phenotypes upon which evolution operates.

In eukaryotic organisms, genetic information is packaged into chromosomes, which carry essential heritable information that must be passed to daughter cells. Mitosis provides a mechanism that ensures each daughter cell receives an identical and complete set of chromosomes and that ensures fidelity in the transmission of heritable information. Mitosis allows for asexual reproduction of organisms in which daughter cells are genetically identical to the parental cell and allows for genetic information transfer to subsequent generations. Both unicellular and multicellular organisms have various mechanisms that increase genetic variation.

Sexual reproduction of diploid organisms involves the recombination of heritable information from both parents through fusion of gametes during fertilization. The two gametes that fuse to form a new progeny zygote each contain a single set (1n) of chromosomes. Meiosis reduces the number of chromosomes from diploid (2n) to haploid (1n) by following a single replication with two divisions. The random assortment of maternal and paternal chromosomes in meiosis and exchanges between sister chromosomes increase genetic variation; thus, the four gametes, while carrying the same number of chromosomes, are genetically unique with respect to individual alleles and allele combinations. The combination of these gametes at fertilization reestablishes the diploid nature of the organism and provides an additional mechanism for generating genetic variation, with every zygote being genetically different. Natural selection operates on populations through the phenotypic differences (traits) that individuals display; meiosis followed by fertilization provides a spectrum of possible phenotypes on which natural selection acts, and variation contributes to the long-term continuation of species.

Some phenotypes are products of action from single genes. These single gene traits provided the experimental system through which Mendel was able to describe a model of inheritance. The processes that chromosomes undergo during meiosis provide a mechanism that accounts for the random distribution of traits, the independence of traits, and the fact that some traits tend to stay together as they are transmitted from parent to offspring. Mendelian genetics can be applied to many phenotypes, including some human genetic disorders. Ethical, social and medical issues can surround such genetic disorders.

Whereas some traits are determined by the actions of single genes, most traits result from the interactions of multiple genes products or interactions between gene products and the environment. These traits often exhibit a spectrum of phenotypic properties that results in a wider range of observable traits, including weight, height and coat color in animals.

Essential knowledge 3.A.1: DNA, and in some cases RNA, is the primary source of heritable information.

a. Genetic information is transmitted from one generation to the next through DNA or RNA.

   Evidence of student learning is a demonstrated understanding of each of the following:

1. Genetic information is stored in and passed to subsequent generations through DNA molecules and, in some cases, RNA molecules.
2. Noneukaryotic organisms have circular chromosomes, while eukaryotic organisms have multiple linear chromosomes, although in biology there are exceptions to this rule.
3. Prokaryotes, viruses and eukaryotes can contain plasmids, which are small extra-chromosomal, double-stranded circular DNA molecules.
4. The proof that DNA is the carrier of genetic information involved a number of important historical experiments. These include:
   i. Contributions of Watson, Crick, Wilkins, and Franklin on the structure of DNA
   ii. Avery-MacLeod-McCarty experiments
   iii. Hershey-Chase experiment
5. DNA replication ensures continuity of hereditary information.
   i. Replication is a semiconservative process; that is, one strand serves as the template for a new, complementary strand.
   ii. Replication requires DNA polymerase plus many other essential cellular enzymes, occurs bidirectionally, and differs in the production of the leading and lagging strands.
6. Genetic information in retroviruses is a special case and has an alternate flow of information: from RNA to DNA, made possible by reverse transcriptase, an enzyme that copies the viral RNA genome into DNA. This DNA integrates into the host genome and becomes transcribed and translated for the assembly of new viral progeny. [See also **3.C.3**]

✘ *The names of the steps and particular enzymes involved, beyond DNA polymerase, ligase, RNA polymerase, helicase and topoisomerase, are outside the scope of the course for the purposes of the AP Exam.*

b. DNA and RNA molecules have structural similarities and differences that define function. [See also **4.A.1**]

Evidence of student learning is a demonstrated understanding of each of the following:

1. Both have three components — sugar, phosphate and a nitrogenous base — which form nucleotide units that are connected by covalent bonds to form a linear molecule with 3' and 5' ends, with the nitrogenous bases perpendicular to the sugar-phosphate backbone.
2. The basic structural differences include:
   i. DNA contains deoxyribose (RNA contains ribose).
   ii. RNA contains uracil in lieu of thymine in DNA.
   iii. DNA is usually double stranded, RNA is usually single stranded.
   iv. The two DNA strands in double-stranded DNA are antiparallel in directionality.
3. Both DNA and RNA exhibit specific nucleotide base pairing that is conserved through evolution: adenine pairs with thymine or uracil (A-T or A-U) and cytosine pairs with guanine (C-G).
   i. Purines (G and A) have a double ring structure.
   ii. Pyrimidines (C, T and U) have a single ring structure.
4. The sequence of the RNA bases, together with the structure of the RNA molecule, determines RNA function.
   i. mRNA carries information from the DNA to the ribosome.
   ii. tRNA molecules bind specific amino acids and allow information in the mRNA to be translated to a linear peptide sequence.
   iii. rRNA molecules are functional building blocks of ribosomes.
   iv. The role of RNAi includes regulation of gene expression at the level of mRNA transcription.

c. Genetic information flows from a sequence of nucleotides in a gene to a sequence of amino acids in a protein.

Evidence of student learning is a demonstrated understanding of each of the following:

1. The enzyme RNA-polymerase reads the DNA molecule in the 3' to 5' direction and synthesizes complementary mRNA molecules that determine the order of amino acids in the polypeptide.
2. In eukaryotic cells the mRNA transcript undergoes a series of enzyme-regulated modifications.

   To foster student understanding of this concept, instructors can choose an illustrative example such as:

   - Addition of a poly-A tail
   - Addition of a GTP cap
   - Excision of introns
3. Translation of the mRNA occurs in the cytoplasm on the ribosome.
4. In prokaryotic organisms, transcription is coupled to translation of the message. Translation involves energy and many steps, including initiation, elongation and termination.

✘ *The details and names of the enzymes and factors involved in each of these steps are beyond the scope of the course and the AP® Exam.*

The salient features include:

i. The mRNA interacts with the rRNA of the ribosome to initiate translation at the (start) codon.

ii. The sequence of nucleotides on the mRNA is read in triplets called codons.

iii. Each codon encodes a specific amino acid, which can be deduced by using a genetic code chart. Many amino acids have more than one codon.

✘ *Memorization of the genetic code is beyond the scope of the course and the AP Exam.*

iv. tRNA brings the correct amino acid to the correct place on the mRNA.

v. The amino acid is transferred to the growing peptide chain.

vi. The process continues along the mRNA until a "stop" codon is reached.

vii. The process terminates by release of the newly synthesized peptide/protein.

d. Phenotypes are determined through protein activities.

To foster student understanding of this concept, instructors can choose an illustrative example such as:

- Enzymatic reactions
- Transport by proteins
- Synthesis
- Degradation

e. Genetic engineering techniques can manipulate the heritable information of DNA and, in special cases, RNA.

To foster student understanding of this concept, instructors can choose an illustrative example such as:

- Electrophoresis
- Plasmid-based transformation
- Restriction enzyme analysis of DNA
- Polymerase Chain Reaction (PCR)

f. *Illustrative examples of products of genetic engineering include:*

- Genetically modified foods
- Transgenic animals
- Cloned animals
- Pharmaceuticals, such as human insulin or factor X