Human Genetics 13th edition Lewis Solutions Manual

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Lewis *Human Genetics*, 13th edition

Answers to selected Key Concepts Questions (that contain *)

Chapter 1

1.1
*b. DNA
*d. Bioethics combines philosophy, science, and medicine to address controversial issues. Bioethics applied to genetics largely concerns privacy.
1.2
*a. Molecules, cells, tissues, organs, organ systems, individuals, families, populations, evolution of species
*c. adenine, cytosine, thymine, guanine
*e. An allele is a variant of a gene.
*f. autosomes and sex chromosomes
*j. A gene pool is all the alleles in a population.
1.3
*a. DNA profiling refers to techniques, statistical tools, and machine learning approaches to compare DNA sequences between individuals, to establish or rule out identity, relationships, or ancestry.
*c. Precision medicine refers to tailoring treatments to a person's gene variants.
*e. Exome sequencing determines the DNA sequences of protein-encoding genes. It can reveal "atypical presentations" of known disorders and diagnose individuals with unusual combinations of symptoms.
1.4
*a. Metagenomics is the study of all of the DNA in a particular geographic or other area.

Chapter 2

2.1
*b. 2
*c. 1
2.2
*a. Eukaryotic cells contain organelles, including a nucleus, and prokaryotic cells do not.
*b. Carbohydrates, lipids, proteins, nucleic acids, vitamins, minerals, and many types of smaller molecules.
*e. The plasma membrane consists of a lipid bilayer with embedded proteins, glycoproteins, and glycolipids. It provides a surface that includes receptors and passageways through which substances (such as signaling molecules and cell adhesion factors) bind or move.
2.3
*b. The cell cycle is divided into interphase, when cellular components are replicated, and mitosis, when contents are distributed into 2 cells.
*c. Mitosis is divided into prophase, metaphase, anaphase, and telophase. Cytokinesis occurs at the end of mitosis and separates the duplicated chromosomes into two daughter cells.
*e. Sister chromatids include a chromosome and an exact copy, held together at the centromere. A replicated chromosome consists of two sister chromatids.
2.4
*b. Self-renewal is the ability of a stem cell to generate one or two daughter cells that are also stem cells.

Chapter 1 The Information in a Human Genome

READINGS

Chapter Opener
Eve's Genome

Clinical Connection 1.1
Genome Sequencing Ends a Child's "Diagnostic Odyssey"

Bioethics: Choices for the Future
Genetic Privacy and Pandora's Box

CHAPTER OVERVIEW

Chapter 1 introduces the basic concepts and language of genetics and genomics, and offers examples of DNA information impacting daily life. People have access to their own genetic information, and health care providers are learning how to incorporate DNA data into diagnosing and threating disease. Bioethics deals with issues of privacy, discrimination, and justice that arise from use and misuse of genetic information. DNA, genes, chromosomes, exomes, and genomes are the levels of genetic information, and they impact biology at the cell, tissue, organ, individual, family, and population levels. Genes encode proteins; the exome is the small part of the genome that does so. Most traits arise from interactions of genes and environmental factors. DNA analysis is useful in studying endangered species, in establishing identity, and in illuminating history. In several nations, large-scale programs are examining exome and genome information, the microbiome, environmental exposures, physiological measurements, and other data to better understand health. Precision medicine attempts to prevent and treat disease based on an individual's gene variants, environmental exposures, and lifestyle factors. Genetic modification has several applications. Metagenomics considers species represented by DNA in the environment.

CHAPTER OUTLINE

1.1 Introducing Genes and Genomes

1. **Genetics** is the study of inherited traits and their variation, and how these traits are passed from one generation to the next (**heredity**).
2. With continuing analysis of human genome sequences, human genetics has grown from a largely academic science to touch many areas of health care, with practical and societal implications. **Genetic genealogy** considers how people are related and where their ancestors lived.
3. **Genes** are the unit of inheritance and are composed of **deoxyribonucleic acid (DNA)**. Genes instruct **cells**, the basic units of life, to manufacture specific proteins.

4. An organism's **genome** is its complete set of genetic information. The **exome** is the portion of the genome that encodes proteins. Sequencing costs have plummeted.
5. **Genomics** is a field of study that reveals how closely related we are to each other and to other species.
6. **Bioethics** addresses issues of privacy, confidentiality, and discrimination that arise from knowledge of our DNA sequences.

1.2 Levels of Genetics and Genomics

Instructions and Information: DNA

1. The levels of genetics and genomics are the molecular (DNA structure and function); cells, tissues, and organs; and families, populations, and species.
2. A DNA molecule consists of "rails" of alternating sugars and phosphates and "steps" of **adenine-thymine** (A-T) and **guanine-cytosine** (G-C) **nitrogenous base** pairs. Each three contiguous base pairs encode one of 20 types of amino acids, which build proteins.
3. DNA copies itself through **replication** and transmits information, while remaining in the nucleus, through **transcription** into RNA.
4. A DNA molecule replicates as the sides of the double helix part and fill in with new bases.
5. After transcription, messenger RNA carries DNA information out of the cell's headquarters (nucleus) to where it is used to synthesize proteins, which is the process of **translation**.
6. The exome is 1.5% of the 20,325 or so genes of the human genome.
7. Genes can exist in more than one form. Variants (**alleles**) arise by **mutation**, which can cause disease. A mutation also refers to a change in a gene.
8. **Chromosomes** consist of hundreds of genes.
9. A human **somatic cell** has 23 pairs of chromosomes, constituting two copies of the genome. 22 pairs are **autosomes** and do not differ between the sexes, and one pair are the **sex chromosomes**. XX is female; XY is male.
10. A **karyotype** is a chart of an individual's chromosomes.
11. A Mendelian trait is caused by a single gene. A **complex trait** is caused by one or more genes and environmental influences. Most genes do not function alone.

The Body: Cells, Tissues, and Organs

1. The human body is composed of about 30 trillion cells. All cell types except mature red blood cells contain two copies of the entire genome.
2. The more than 290 specialized, or **differentiated,** cell types express different subsets of genes. Differentiated cells interact, forming four basic **tissue** types that interact, forming organs and organ systems.
3. Many organs contain **stem cells** that "self-renew" and produce differentiated cells, properties that are essential for growth, development, and healing.

Relationships: From Individuals to Families

1. **Genotype** is the allelic makeup of an individual; **phenotype** is the observable or measureable expression of an individual's alleles (traits or health condition).
2. **Dominant** alleles are expressed when one copy is present. **Recessive** alleles require two copies for expression.

3. **Pedigrees** are diagrams that depict the transmission of recessive and dominant traits through generations. The proportion of the genome that an individual shares with an ancestor halves at each generation.

The Bigger Picture: From Populations to Evolution

1. Population genetics concerns allele frequencies in members of the same species in a specified geographic area.
2. A **gene pool** is all of the alleles in a given population.
3. Population genetics has applications in health care and forensics and underlies the changes of evolution.
4. The more closely two individuals' DNA sequences match, the more recently they shared an ancestor.
5. Comparative genomics explores evolutionary relationships among species.

1.3 Applications of Genetics and Genomics

Establishing Identity

1. **DNA profiling** compares DNA sequences among individuals. The more DNA sequences individuals share, the more closely related they are and the more recently they've shared ancestors.
2. It is used to establish or rule out identity, clarify relationships or ancestry, and to evaluate crime scenes, probe sites of natural disasters, reunite adopted individuals with birth parents, and test food for contamination or mislabeling.

Illuminating History

1. DNA analysis can illuminate history by confirming relationships of people to each other and determining where people came from.
2. DNA testing provides information on past epidemics and reveals genetic diversity of a population, indicating potential need for species conservation efforts.

Precision Medicine

1. Large projects compare many types of information to learn about health, including genome sequences, the microbes on and in the body (the **microbiome**), environmental exposures, diet, and activity levels.
2. **Pharmacogenetics** predicts responses of individuals to drugs based on genotypes.
3. Healthy and long-lived individuals provide valuable information on the genes that keep us well.

Genetic Modification

1. Genetic modification alters a gene or genome in a way that does not occur in nature, such as combining DNA sequences from individuals of different species.
2. Genetically modified organisms include bacteria that produce human proteins used as drugs and altered crop plants.

3. **Genome editing** adds, deletes, or replaces specific genes.

Exome Sequencing

1. Exome sequencing can lead to diagnoses of conditions that are unrecognized from their symptoms by implicating specific DNA sequences.

2. Exome and genome sequencing can shorten the time it takes to diagnose a rare disease, sometimes from years to days.

1.4 A Global Perspective on Genomes

1. **Metagenomics** considers sequences of all DNA in a habitat, from an ocean to a small body part.
2. Social issues that arise from genetic and genomic technologies include access to, misuse of, and abuse of DNA information.
3. Nations are individualizing guidelines to maximize benefit from emerging genetic tests and technologies.

IDEAS FOR DISCUSSION

1. Bioethics is a field of study concerned with issues of privacy, confidentiality, and discrimination that arise from access to DNA sequences and their interpretation. The **Bioethics** essay at the end of chapter 1 addresses privacy issues with collecting personal genetic information in certain situations and scenarios. How do students feel about access to their own genetic information? Does a parent have an obligation to inform a child of a possible mutation, and if so, under what circumstances? Should an employer offer health insurance on the contingency that an employee have genetic testing? How can genetic testing affect people other than the one who is tested?

2. Students today learn about DNA in grade school. However, coverage of genetics in the media often deals with cases and emotional issues, rather than the science. Have students discuss how information is part of the DNA molecule. How does a DNA polymer differ from a carbohydrate or lipid in its information content?

3. Which genes or inherited traits or conditions would students want to know about in their future offspring, and which would they not want to know about? Contrast advantages and dangers of knowing such information. What should be done if parents disagree?

4. Millions of people have taken consumer DNA tests, as Figure 1.1 illustrates. Sometimes the results of these tests are not what people expected, and can be upsetting. The author of the textbook discovered recently that she was conceived from a sperm donor and has several half-siblings. In her family and many others, younger people take the tests, and the findings reveal events that happened in the lives of their much older relatives. Should a person who has not taken a consumer DNA test nevertheless be informed if someone else's test alters her or his view of the place in the family? How should these tests be used?

TIMELY ARTICLES BY RICKI LEWIS

The author writes weekly articles on genetics in the news. Access posts at her blog *DNA Science* (hosted by Public Library of Science) at http://blogs.plos.org/dnascience/author/rlewis/ and her posts at *Genetic Literacy Project* at https://geneticliteracyproject.org/tag/ricki-lewis/. She reposts these at www.rickilewis.com/blog and at her Amazon author page https://www.amazon.com/Ricki-Lewis/e/B001IO9RK6.

ANSWERS TO REVIEW QUESTIONS

1. Gene pool, genome, chromosome, gene, DNA

2. A bioethical issue would be an employer requesting information about whether or not an employee has inherited a susceptibility gene for breast cancer, which might affect relatives of the employee.

3. The sequence of DNA nucleotides (A, G, C, T) in a gene comprises a genetic code that is read three nucleotides at a time to direct the building of proteins from amino acids.

4. a. An autosome does not carry genes that determine sex. A sex chromosome does.
   b. Genotype is the allele constitution in an individual for a particular gene. Phenotype is the physical expression of an allele combination.
   c. DNA is a double-stranded nucleic acid that includes deoxyribose and the nitrogenous bases adenine, guanine, cytosine, and thymine. DNA carries the genetic information. RNA is a single-stranded nucleic acid that includes ribose and the nitrogenous bases adenine, guanine, cytosine, and uracil. RNA carries out gene expression.
   d. A recessive allele determines phenotype in two copies. A dominant allele determines phenotype in one copy.
   e. A pedigree is a chart of family relationships and traits. A karyotype is a chart of chromosomes.
   f. A gene is a sequence of DNA that encodes a protein. A genome is all DNA in a set of genetic instructions. Most human cells have two copies of the genome.
   g. An exome is the protein-encoding part of a genome. A genome is all the DNA in a set of genetic instructions.
   h. A Mendelian trait comes from a single gene, whereas a complex trait arises from genes and environmental factors.

5. An allele is a gene variant.

6. Differential gene expression creates distinctive cell types.

7. Pedigrees depict traits or illnesses and how people are related to one another.

8. A gene pool is the collection of alleles in a population.

9. A use of determining a DNA sequence is to diagnose a disease or identify a criminal. DNA manipulation is used to create crops with a desired trait.

10. Metagenomics analyzes all of the DNA in an area, such as microbes on a subway railing, and the people, dogs, rodents, and insects in the subway station.

ANSWERS TO APPLIED QUESTIONS

1. A risk of making a healthcare decision based on a consumer genetic test is that the test may detect only the most common gene variants. A benefit is that detecting a mutation can alert a person to a health risk, such as detecting a cancer risk gene variant.

2. People may not realize that the drugs that they take are produced in bacteria that use genes from humans. The media focuses more on GMOs that we eat than the ones that provide drugs.

3. Carrying genetic information on a smartphone might be lifesaving if the information is medically important. Or it can tell too much about a person as well as relatives.

4. A frivolous use of DNA testing is to discover hair or eye color. A serious use of DNA testing is to diagnose an illness.

5. Answers will vary.

ANSWERS TO FORENSICS FOCUS

1. DNA testing might distinguish DNA from Romeo, seafood in the cat food, liver in the treats, eggplant from the counter, whatever is on his fur and whatever he ate outdoors.

2. Genetic information might be useful in a legal case if a particular genotype can account for a very specific behavior that caused a person to commit a crime.

3. DNA can be collected from cheek swabs from people separated who may be related, and sets of DNA markers compared. The proportion of shared markers reveals the relationship. Full siblings would share half, parents and children half, half-siblings, a quarter of their DNA.

ANSWERS TO CASE STUDIES AND RESEARCH RESULTS

1. Sequencing genomes of deceased individuals might reveal whether they were correctly diagnosed and treated, as well as missed diagnoses.

2. Some people might be upset to see their image displayed in a gallery, the piece derived from analysis of their DNA collected from a discarded object. But in this day and age when people photograph anyone they want with their phones, one could argue that privacy no longer exists.

3. Parents might want to know what exactly will be done with their children's DNA at the time of collection and going forward; who will have access to the information; and how the information will be protected.

ANSWERS TO CLINICAL CONNECTION 1.1 QUESTIONS

1. Millie was a new mutation, so her siblings would not be at risk.

2. Yes

3. A parent often feels better knowing a diagnosis because information is then available about future symptoms and problems. It is also helpful for when a treatment is developed or an existing drug repurposed. A diagnosis also enables parents to find other affected families who can provide helpful advice and resources.

ANSWERS TO BIOETHICS

1. People objected to the Berkeley student testing because students could have felt coerced to participate.
2. Opinion on when a government should require DNA testing. Possibilities include for arrested individuals, to reunite separated families, to diagnose genetic disease in newborns.
3. A student taking a DNA test in a class situation might be alerted to a health condition early enough for treatment following a clinical test from a healthcare professional. A student taking an ancestry test may discover a geographic background at odds with previous knowledge.
4. The answer depends upon circumstance. A student discovering half-siblings who knew she was donor conceived, for example, might have a very different reaction from a student finding out about having half-siblings who did not know the circumstances of conception.

ANSWERS TO KEY CONCEPTS QUESTIONS

1.1

1. a. Genetics is the study of how traits are transmitted. Heredity considers the transmission patterns of inherited traits between generations.
2. *b. DNA
3. c. A gene is a sequence of DNA that encodes a protein. An exome consists of protein-encoding genes. A genome is a complete set of genetic instructions for an organism.
4. *d. Bioethics combines philosophy, science, and medicine to address controversial issues. Bioethics applied to genetics largely concerns privacy.

1.2

1. *a. Molecules, cells, tissues, organs, organ systems, individuals, families, populations, evolution of species
2. b. DNA carries information in the sequence of nitrogenous bases (A, C, T and G)
3. *c. adenine, cytosine, thymine, guanine
4. d. A mutation can cause a disease by altering the amino acid sequence of a protein.
5. *e. An allele is a variant of a gene.

6. *f. autosomes and sex chromosomes
7. g. A Mendelian trait arises from inheriting a mutation in one gene. A complex trait derives from variants of one or more genes plus environmental influences.
8. h. Gene expression refers to transcription of RNA from the DNA of a gene. Cells specialize by expressing subsets of protein-encoding genes in the genome.
9. i. Genotype is an allele combination; phenotype is the expression of an allele combination (trait or health condition). A dominant allele affects the phenotype in one copy. A recessive allele affects the phenotype in two copies.
10. *j. A gene pool is all the alleles in a population.
11. k. The more recently two species shared an ancestor (are more closely related), the more of their DNA sequence they share.

1.3

1. *a. DNA profiling refers to techniques, statistical tools, and machine learning approaches to compare DNA sequences between individuals, to establish or rule out identity, relationships, or ancestry.
2. b. DNA profiling can establish if and how individuals are related to one another.
3. *c. Precision medicine refers to tailoring treatments to a person's gene variants.
4. d. Traditional breeding combines existing traits in novel ways. Genetic modification alters DNA sequences
5. *e. Exome sequencing determines the DNA sequences of protein-encoding genes. It can reveal "atypical presentations" of known disorders and diagnose individuals with unusual combinations of symptoms.

1.4

1. *a. Metagenomics is the study of all of the DNA in a particular geographic or other area.
2. b. Access to genetic testing and prioritizing testing for genetic diseases when other types of illnesses, such as infections, are more prevalent.

ADDITIONAL QUESTIONS

1. Osteoporosis thins bones in millions of people. A much rarer, inherited condition, osteopetrosis, acts oppositely, increasing bone mass. Osteopetrosis does not affect health, and is typically discovered on x-rays. Why might studying how osteopetrosis arises be useful, even though it doesn't cause symptoms?

2. In acute intermittent porphyria, an environmental factor such as abuse of drugs or alcohol, or fasting, triggers potentially fatal attacks on the nervous system. An abnormal enzyme causes the condition. Why is this disorder considered genetic if it only produces symptoms in the presence of a specific environmental trigger?

3. Although seizures are usually not inherited, a condition called benign infantile familial convulsions run in families. How can studying this inherited illness provide information that may help people with noninherited seizures?

4. Due to inherited differences in the way the body processes cholesterol, some people can eat a fatty diet yet have healthy blood serum cholesterol, and others develop dangerously high cholesterol levels if they do not eat wisely. Several drugs can lower blood serum cholesterol level. Researchers found that people with a certain variant of a gene encoding a protein that transports cholesterol into liver cells (cholesteryl ester transfer protein, or CETP) are likely to have a cholesterol problem, and are also more likely to benefit from cholesterol-lowering drugs than people with different alleles. What information would be important to have before undergoing a CETP gene test and possibly taking a cholesterol-lowering drug?

5. In Graves disease, the immune system attacks the thyroid gland, which normally produces hormones controlling energy utilization. Siblings of people with Graves disease are 15 times as likely to develop the disorder as people whose siblings are unaffected. Women develop the condition more often than men, and a high percentage of affected individuals smoke. Is Graves disease is caused solely by an

abnormal gene, solely by an environmental trigger, or might there be another explanation?

ANSWERS TO ADDITIONAL QUESTIONS

1. Studying osteopetrosis can reveal how healthy bone mass is maintained, which might provide insights into osteoporosis, which has an opposite phenotype and is more common. Drugs may be developed that mimic osteopetrosis to treat osteoporosis.

2. The condition is inherited because presence of a particular gene variant is necessary for symptoms to occur. The susceptibility is inherited, not the disease.

3. Studying this rare inherited condition can reveal basic brain mechanisms that may explain how more common seizures occur.

4. Important information includes: the risk of cardiovascular disease to an individual; side effects of cholesterol-lowering drugs; the role of diet in controlling gene expression; how often a particular genetic variant is associated with increased cholesterol level; success of drug treatment.

5. Graves disease might be caused by an inherited susceptibility triggered by an environmental influence, such as exposure to female sex hormones or cigarette smoke, or the status of the immune system.