Human Genetics 14th edition Lewis Solutions Manual

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Chapter 1 The Information in a Human Genome

Chapter Opener

Eve's Genome

Bioethics

Genetic Privacy and Pandora's Box

CHAPTER OVERVIEW

Chapter 1 introduces the basic concepts and language of genetics and genomics, with eclectic examples of how DNA information can impact daily life. People have access to their own genetic information, and health care providers are learning how to incorporate DNA data into diagnosing and treating disease. DNA, genes, chromosomes, exomes, and genomes are levels of genetic information. 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. Human genomes vary. Most traits arise from interactions of genes and environmental factors. DNA analysis is useful in establishing identity and in illuminating history. Several nations store genetic information of citizens in population biobanks. Precision medicine strives 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. The bioethics box addresses issues of privacy, discrimination, and justice that arise from use and misuse of genetic information.

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. Most genes are in a cell's **nucleus**.
4. A **genome** is an organism's complete set of genetic information. The **exome** is the portion of the genome that encodes proteins.
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

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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 new bases.
5. After transcription, messenger RNA carries DNA information out of the nucleus to the cytoplasm, where it is used to synthesize proteins, which is called **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 changes the DNA base sequence. Many studies compare thousands of places in the genome that vary among populations.
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. 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 single gene causes a Mendelian trait. One or more genes and environmental influences cause a **complex trait**. 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 form organs and organ systems.
3. **Stem cells** "self-renew" and produce differentiated cells. Self-renewal is 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.

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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 families, and test food for contamination or mislabeling.

Illuminating History

1. DNA analysis can confirm relationships among individuals and determine where people came from.
2. DNA testing provides information on past epidemics and reveals genetic diversity of populations.

Biobanks

1. A **biobank** stores biological samples or DNA sequence data and information on environmental exposures and lifestyle habits.
2. The largest biobank is the UK Biobank.
3. Biobanks are making efforts to more accurately reflect the diversity of humanity.

Precision Medicine

1. A precision medicine approach uses DNA data to select treatments for individuals most likely to be effective and least likely to have intolerable adverse effects.
2. **Pharmacogenetics** predicts responses of individuals to drugs based on genotypes.

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 (**recombinant DNA technology**).
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 and Genome Sequencing

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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. This is environmental (or e) DNA.
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 considers issues of privacy, confidentiality, and discrimination that arise from access to DNA sequences and their interpretation. The **Bioethics** essay addresses privacy issues that can arise from 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 only to employees who agree to have genetic testing? How can genetic testing affect people other than the individual 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. The first vaccines to protect against COVID-19 consisted of messenger RNA. Some people who refused to be vaccinated did so because they thought that the RNA would change their DNA. Discuss how these vaccines work (see figure 10B) and discuss how people can be educated about the relationship of DNA, RNA, and protein. How can vaccination be viewed from a personal and a population perspective?

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

5. Millions of people have taken consumer DNA tests, as Figure 1.1 illustrates. Results of these tests may not be what people expected, and can be upsetting. The author of the textbook discovered that she was conceived from a sperm donor and has several half-siblings, from a DNA test that she took at a genetics conference. 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

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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) distinguishes individuals

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 a phenotype in two copies. A dominant allele determines a 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.

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

ANSWERS TO CRITICAL THINKING QUESTIONS

1. A risk of making a healthcare decision based on a consumer DNA 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 a cancer risk gene variant.

2. 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.

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

4. Answers will vary.

5. mRNA holds the code for an amino acid sequence of a specific protein.

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, what is on his fur and what 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, deduced from discarded DNA collected without permission.

3. Parents might want to know the intended use of their children's DNA information, access to it, and how the information and the child's identity will be protected.

4. UK Biobank studies include diet and cardiovascular disease links considering genetic background, severity of COVID-19, gene variants behind rare diseases, gene variants that predispose to dementia, gout, polygenic risk scores for many common illnesses, connections among common gene variants and behavioral disorders.

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5. Exercise and activity, stress, economic status

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

CHAPTER 1

1.1
a. Genetics is the study of how traits are transmitted. Heredity considers transmission patterns of inherited traits between generations.
b. DNA
c. A gene is a sequence of DNA that encodes a protein. An exome is protein-encoding genes. A genome is a complete set of genetic instructions for an organism.
d. Bioethics combines philosophy, science, and medicine to address controversial issues. Bioethics applied to genetics largely concerns privacy and manipulations.

1.2
a. Molecules, cells, tissues, organs, organ systems, individuals, families, populations, evolution of species
b. DNA carries information in the sequence of nitrogenous bases (A, C, T and G)
c. adenine, cytosine, thymine, guanine
d. A mutation can cause disease by altering the amino acid sequence of a protein.
e. An allele is a variant of a gene.
f. autosomes and sex chromosomes
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.
h. Gene expression refers to transcription of RNA from the DNA of a gene. Cells specialize by expressing subsets of protein-encoding genes.
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.
j. A gene pool is all the alleles in a population.
k. The more recently two species shared an ancestor (are more closely related) the more of their DNA sequence they share.

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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.
b. DNA profiling can determine if and how individuals are related to one another, including identifying descendants of people who were prominent in history.
c. Biobank data can be used to identify environmental and/or genetic causes of disease, which can inform development of new treatments and to identify existing drugs most likely to work for an individual (pharmacogenetics).
d. Precision medicine refers to tailoring treatments to a person's gene variants.
e. Traditional breeding combines existing traits in novel ways. Genetic modification alters DNA sequences.
f. Exome and genome sequencing more precisely diagnose illnesses from the DNA sequences of protein-encoding genes. They can speed diagnoses of rare conditions.

1.4
a. Metagenomics is the study of all DNA in a geographic or other area.
b. Social issues include 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 more rare, 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 fasting or drinking alcohol excessively, 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 runs 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 caused solely by an

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

6. Give an example of how metagenomics was used in understanding the cause of COVID-19.

7. Discuss why the phrase “the human genome” is inaccurate.

8. Describe a use of data from a biobank.

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.

6. Identifying RNA from the virus SARS-CoV-2 in certain non-human organisms such as deer, on surfaces, and in parts of the human body.

7. Human genomes vary.

8. Biobank data can help to reveal gene variants and environmental exposures associated with a subtype of a disease or with patients who have a poor prognosis.

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