SOLUTIONS AND PROBLEM-SOLVING MANUAL
FOR

# *Genetics Essentials:*
## *Concepts and Connections*

Third Edition

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# Chapter One: Introduction to Genetics

## COMPREHENSION QUESTIONS

### Section 1.1

*1. How did the Hopi culture contribute to the high incidence of albinism among members of the Hopi tribe?

Solution:
In the Hopi culture, albino individuals were considered special and awarded special status in the village. Hopi male albinos were not required to work the fields, thus avoiding extensive exposure to sunlight that could prove damaging or deadly. Albinism was considered a positive trait and not a negative physical condition, which allowed albinos to have more children, increasing the frequency of the albino allele. Finally, the small population size of the Hopi tribe may have helped increase the allele frequency of the albino gene due to chance.

2. Give at least three examples of the role of genetics in society today.

Solution:
Genetics plays important roles in the diagnosis and treatment of hereditary diseases; in breeding plants and animals for improved production and disease resistance; and in producing pharmaceuticals and novel crops through genetic engineering.

3. Briefly explain why genetics is crucial to modern biology.

Solution:
Genetics is crucial to modern biology in that it provides unifying principles: all organisms use nucleic acid as their genetic material, and all organisms encode genetic information in the same manner. The study of many other biological disciplines, such as developmental biology, ecology, and evolutionary biology, is supported by genetics.

4. List the three traditional subdisciplines of genetics and summarize what each covers.

Solution:
Transmission genetics: inheritance of genes from one generation to the next, gene-mapping.
Molecular genetics: structure, organization, and function of genes at a molecular level.
Population genetics: the genetic composition of populations and how the genetic composition changes over time.

5. What are some characteristics of model genetic organisms that make them useful for genetic studies?

Solution:

Model genetic organisms have relatively short generation times, produce numerous progeny, are amenable to laboratory manipulations, and can be maintained and propagated inexpensively.

**Section 1.2**

6. When and where did agriculture first arise? What role did genetics play in the development of the first domesticated plants and animals?

Solution:
Agriculture first arose 10,000 to 12,000 years ago in the area now referred to as the Middle East (i.e., Turkey, Iran, Iraq, Syria, Jordan, and Israel). Early farmers selectively bred individual wild plants or animals that had useful characteristics with others that had similar useful traits. The farmers then selected for offspring that contained those useful features. Early farmers did not completely understand genetics, but they clearly understood that breeding individual plants or animals with desirable traits would lead to offspring that contained these same traits. This selective breeding led to the development of domesticated plants and animals.

7. Outline the concept of pangenesis and explain how it differs from the germ-plasm theory.

Solution:
Pangenesis theorizes that information for creating each part of the offspring's body originates from each part of the parent's body and is passed through the reproductive organs to the embryo at conception. Pangenesis suggests that changes in parts of the parent's body may be passed to the offspring's body. The germ-plasm theory, in contrast, states that the reproductive cells possess all of the information required to make the complete body; the rest of the body contributes no information to the next generation.

8. What does the concept of the inheritance of acquired characteristics propose and how is it related to the notion of pangenesis?

Solution:
The theory of inheritance of acquired characteristics postulates that traits acquired during one's lifetime can be transmitted to offspring. It developed from pangenesis, which postulates that information from all parts of one's body is transmitted to the next generation. Thus, for example, learning acquired in the brain or larger arm muscles developed through exercise could be transmitted to offspring.

9. What is preformationism? What did it have to say about how traits are inherited?

Solution:
Preformationism is the theory that the offspring results from a miniature adult form already preformed in the sperm or the egg. All traits would thus be inherited from only one parent, either the father or the mother, depending on whether the homunculus (the preformed miniature adult) resided in the sperm or the egg.

10. Define blending inheritance and contrast it with preformationism.

    Solution:
    The theory of blending inheritance proposes that the egg and sperm from two parents contain material that blends upon conception, influencing the development of the offspring. This theory indicates that the offspring is an equal blend of the two parents. In preformationism, the offspring inherits all of its traits from one parent.

11. How did developments in botany during the seventeenth and eighteenth centuries contribute to the rise of modern genetics?

    Solution:
    Botanists of the seventeenth and eighteenth centuries developed new techniques for crossing plants and creating plant hybrids. These early experiments provided essential background work for Mendel's plant crosses. Mendel's work laid the foundation for the study of modern genetics.

12. List some advances in genetics made in the twentieth century.

    Solution:
    1902 Proposal that genes are located on chromosomes by Walter Sutton
    1910 Discovery of the first genetic mutation in a fruit fly by Thomas Hunt Morgan
    1930 The foundation of population genetics by Ronald A. Fisher, John B. S. Haldane, and Sewall Wright
    1940s The use of viral and bacterial genetic systems
    1953 Three-dimensional structure of DNA described by Watson and Crick
    1966 Deciphering of the genetic code
    1973 Recombinant DNA experiments
    1977 Chemical and enzymatic methods for DNA sequencing developed by Walter Gilbert and Frederick Sanger
    1986 PCR developed by Kary Mullis
    1990 Gene therapy

13. Briefly explain the contribution that each of the following persons made to the study of genetics.

    Solution:

    **a**. Matthias Schleiden and Theodor Schwann

    Proposed the concept of the cell theory, which indicated that the cell is the fundamental unit of living organisms. Caused biologists interested in heredity to examine cell reproduction.

    **b.** August Weismann

    Proposed the germ-plasm theory, which holds that cells in reproductive organs carry a complete set of genetic information.

    **d**. Gregor Mendel

First discovered the basic rules of inheritance.

**e**. James Watson and Francis Crick

Along with Roslind Franklin and Maurice Wilkins, described the three-dimensional structure of DNA.

**f**. Kary Mullis

Developed the polymerase chain reaction, used to quickly amplify small amounts of DNA.

**Section 1.3**

14. What are the two basic cell types (from a structural perspective) and how do they differ?

Solution:
The two basic cell types are prokaryotic and eukaryotic. Prokaryotic cells do not have a nucleus, and their chromosomes are found within the cytoplasm. They do not possess membrane-bound cell organelles. Eukaryotic cells possess a nucleus and membrane-bound cell organelles.

*15. Summarize the relations between genes, DNA, and chromosomes.

Solution:
Genes are composed of DNA nucleotide sequences that are located at specific positions in chromosomes.

## APPLICATION QUESTIONS AND PROBLEMS

**Section 1.1**

*16. How are genetics and evolution related?

Solution:
Evolution is genetic change over time. For evolution to occur, genetic variation must first arise, and then evolutionary forces change the proportion of genetic variants over time. Genetic variation is therefore the basis of all evolutionary change.

*17. For each of the following genetic topics, indicate whether it focuses on transmission genetics, molecular genetics, or population genetics.

**a.** Analysis of pedigrees to determine the probability of someone inheriting a trait

Solution: Transmission genetics

**b.** Study of people on a small island to determine why a genetic form of asthma is so prevalent on the island

Solution: Population genetics

**c.** Effect of nonrandom mating on the distribution of genotypes among a group of animals

Solution: Population genetics

**d.** Examination of the nucleotide sequences found at the ends of chromosomes

Solution: Molecular genetics

**e.** Mechanisms that ensure a high degree of accuracy during DNA replication

Solution: Molecular genetics

**f.** Study of how the inheritance of traits encoded by genes on sex chromosomes (sex-linked traits) differs from the inheritance of traits encoded by genes on nonsex chromosomes (autosomal traits)

Solution: Transmission genetics

**Section 1.2**

*18. Genetics is said to be both a very old science and a very young science. Explain what this means.

Solution:
Genetics is old in the sense that humans have been aware of hereditary principles for thousands of years and have applied these principles since the beginning of agriculture. It is very young in the sense that the fundamental principles were not uncovered until Mendel's time, and the discovery of the structure of DNA and the principles of recombinant DNA have occurred within the last 60 years.

*19. Match each description (*a* through *d*) with the correct theory or concept listed below.

Preformationism

Pangenesis

Germ-plasm theory

Inheritance of acquired characteristics

**a.** Each reproductive cell contains a complete set of genetic information.

Solution: Germ-plasm theory

**b.** All traits are inherited from one parent.

Solution: Preformationism

**c.** Genetic information may be altered by the use of a characteristic.

Solution: Inheritance of acquired characteristics

**d.** Cells of different tissues contain different genetic information.

Solution: Pangenesis

*20. Compare and contrast the following ideas about inheritance.

**a.** Pangenesis and germ-plasm theory

Solution:
Pangenesis postulates that particles carry genetic information from all parts of the body to the reproductive organs, and then the genetic information is conveyed to the embryo, where each unit directs the formation of its own specific part of the body. According to the germ-plasm theory, gamete-producing cells found within the reproductive organs contain the complete set of genetic information that is passed to the gametes. Pangenesis and the germ-plasm theory are similar in that both propose that genetic information is contained in discrete units that are passed on to the offspring. They differ in where that genetic information resides. In pangenesis, it resides in different parts of the body and must travel to the reproductive organs. In the germ-plasm theory, all the genetic information is already in the reproductive cells.

**b.** Preformationism and blending inheritance

Solution:
Preformationism holds that the sperm or egg contains a miniature preformed adult called the homunculus. In development, the homunculus grows to produce an offspring. Only one parent contributes genetic traits to the offspring. Blending inheritance requires contributions of genetic material from both parents. The genetic contributions from the parents blend to produce the genetic material of the offspring. Having been blended, the genetic material cannot be separated for future generations.

**c.** The inheritance of acquired characteristics and our modern theory of heredity

Solution:
The inheritance of acquired characteristics postulates that traits acquired in a person's lifetime alter the genetic material and can be transmitted to offspring. Our modern theory of heredity indicates that offspring inherit genes located on chromosomes passed from their parents. These chromosomes segregate in meiosis in the parent's germ cells and are passed into the gametes.

**Section 1.3**

*21. Compare and contrast the following terms:

**a.** Eukaryotic and prokaryotic cells

Solution:
Both cell types have lipid bilayer membranes, DNA genomes, and machinery for DNA replication, transcription, translation, energy metabolism, response to stimuli, growth, and reproduction. Eukaryotic cells have a nucleus containing chromosomal DNA and possess internal membrane-bound organelles.

**b.** Gene and allele

Solution:
A gene is a basic unit of hereditary information, usually encoding a functional RNA or polypeptide. Alleles are variant forms of a gene, arising through mutation.

**c.** Genotype and phenotype

Solution:
The genotype is the set of genes or alleles inherited by an organism from its parent(s). The expression of the genes of a particular genotype, through interaction with environmental factors, produces the phenotype, the observable trait.

**d.** DNA and RNA

Solution:
Both are nucleic acid polymers. RNA contains a ribose sugar, whereas DNA contains deoxyribose sugar. RNA also contains uracil as one of the four bases, whereas DNA contains thymine. The other three bases are common to both DNA and RNA. Finally, DNA is usually double-stranded, consisting of two complementary strands, whereas RNA is single-stranded

**e.** DNA and chromosome

Solution:
Chromosomes are structures consisting of DNA and associated proteins. The DNA contains the genetic information.

## CHALLENGE QUESTIONS

**Introduction**

*22. The type of albinism that arises with high frequency among the Hopis (discussed in the introduction to this chapter) is most likely oculocutaneous albinism type 2, which is caused by a defect in the *OCA2* gene on chromosome 15. Do some research on the Internet to determine how the phenotype of this type of albinism differs from phenotypes of other forms of albinism in humans and the mutated genes that result in these phenotypes. Hint: Visit the website Online Mendelian Inheritance in Man and search the database for albinism.

Solution:

| **Type of albinism** | **Phenotype** | **Gene mutated** |
|---|---|---|
| OCA2 | Pigment reduced in skin, hair, and eyes, but small amount of pigment acquired with age; visual problems | *OCA2* |
| OCA1B | General absence of pigment in hair, skin, and eyes, but there may be small amount of pigment; does not vary with age; visual problems | Gene that encodes tyrosinase |
| OCA1A | Complete absence of pigment; visual problems | Gene that encodes tyrosinase |
| OCA3 | Some pigment present; sun sensitivity and visual problems | Gene that encodes tyrosinase-related protein 1 |
| OASD | Lack of pigment in the eyes and deafness later in life | Unknown |
| OA1 | Lack of pigment in the eyes but normal elsewhere | *GPR143* |
| ROCA | Bright copper-red coloration in skin and hair of Africans; dilution of color in iris | Gene that encodes tyrosinase-related protein 1 |
| OCA4 | Reduced pigmentation | *MATP* |

**Section 1.1**

23. We now know a great deal about the genetics of humans. What are some of the reasons humans have been the focus of intensive genetic study?

Solution:
Humans are intensively interested in how humans function biologically. Because of this intense interest, we know more about the anatomy, physiology, genetics, and biochemistry of humans than of many other organisms. Many human diseases and disorders are associated with human genes. Understanding how to treat and diagnose these diseases and

disorders requires intensive studies to identify the gene(s) responsible for the disorder as well as understanding how they are inherited and expressed. Recent advances in the understanding of genetic risk factors associated with diseases such as heart disease and cancer have enabled the development of predictive genetic tests for some of these disorders. These successes continue to stimulate a focus in identifying genetic risk factors for other diseases. The ability of families to keep careful records about members extending back many generations has facilitated the study of human inheritance that has aided the ability of researchers to identify genetic markers within families. In addition, these detailed records have provided some humans who are intensely interested in their own heredity the ability to trace their ancestry.

24. Describe some of the ways in which your own genetic makeup affects you as a person. Be as specific as you can.

Solution:
Answers will vary but should include observations similar to those in the following example: Genes affect my physical appearance; for example, they probably have largely determined the fact that I have brown hair and brown eyes. Undoubtedly, genes have affected my height of five feet, seven inches, which is quite close to the height of my father and mother, and my slim build. My dark complexion mirrors the skin color of my mother. I have inherited susceptibilities to certain diseases and disorders that tend to run in my family; these include asthma, a slight tremor of the hand, and vertigo.

25. Describe at least one trait that appears to run in your family (appears in multiple members of the family). Do you think that this trait runs in your family because it is an inherited trait or because it is caused by environmental factors that are common to family members? How might you distinguish between these possibilities?

Solution:
My two brothers and I share two traits: all three of us are both taciturn (we don't speak much) and smart (just don't ask my teenage daughter). Although the literature provides evidence for a genetic component for intelligence, I'm not aware of any studies on the heritability of being taciturn. If I were to investigate the extent to which these traits are determined by the environment or by heredity, I would look at studies of twins who had been separated at birth and lived in different environments to adulthood. Such studies would separate environmental factors from genetic factors, whereas studies of family members reared in the same household are confounded by the fact that the family members experienced similar environments. If the trait had a strong genetic component, we would expect identical twins reared apart to be similarly taciturn or similarly intelligent. One would have to devise some objective measure of these traits–degrees of being taciturn or smart.

**Section 1.3**

*26. Suppose that life exists elsewhere in the universe. All life must contain some type of genetic information, but alien genomes might not consist of nucleic acids and have the same features as those found in the genomes of life on Earth. What do you think might be the common features of all genomes, no matter where they exist?

Solution:
All genomes must have the ability to store complex information and to vary. The blueprint for the entire organism must be contained within the genome of each reproductive cell. The information has to be in the form of a code that can be used as a set of instructions for assembling the components of the cells. The genetic material of any organism must be stable, be replicated precisely, and be transmitted faithfully to the progeny but must be capable of mutating.

27. Choose one of the ethical or social issues in parts *a* through *d* and give your opinion on the issue. For background information, you might read one of the articles on ethics listed and marked with an asterisk in the Suggested Readings section for Chapter 1 in your LaunchPad.

**a.** Should a person's genetic makeup be used in determining his or her eligibility for life insurance?

Solution:
**Arguments pro:** Genetic susceptibility to certain types of diseases or conditions is relevant information regarding consequences of exposure to certain occupational hazards. Genes that will result in neurodegenerative diseases, such as Huntington disease, Alzheimer disease, or breast cancer, could logically be considered preexisting conditions. Insurance companies have a right, and arguably a duty to their customers, to exclude people with genetic preconditions so that insurance rates can be lowered for the general population.
**Arguments con:** The whole idea of insurance is to spread the risk and pool assets. Excluding people based on their genetic makeup would deny insurance to people who need it most. Indeed, as information about various genetic risks accumulates, more people would become excluded until only a small fraction of the population is insurable.

**b.** Should biotechnology companies be able to patent newly sequenced genes?

Solution:
**Pro:** Patenting genes provides companies with protection for their investment in research and development of new drugs and therapies. Without such patent protection, companies would have less incentive to expend large amounts of money in genetic research, which would slow the pace of advancement of medical research. Such a result would be detrimental to everyone.
**Con:** Patents on human genes would be like allowing companies to patent a human arm. Genes are integral parts of our selves, so how can a company patent something that every human has?

**c.** Should gene therapy be used on people?

Solution:
**Pro:** Gene therapy can be used to cure previously incurable or intractable genetic disorders and to relieve the suffering of millions of people.
**Con:** Gene therapy may lead to genetic engineering of people for unsavory ends. Who determines what is a genetic defect? For example, would short stature be considered a genetic defect?

**d.** Should genetic testing for inherited disorders for which there is no treatment or cure be made available?

Solution:
**Pro:** Information will provide relief from unnecessary anxiety (if the test is negative). Even if the test result is positive for a genetic disorder, it provides the individual, the family, and friends with information and time to prepare. Information about one's own genetic makeup is a right; every person should be able to make his or her own choice as to whether he or she wants this information.
**Con:** If there is no treatment or cure, a positive test result can have no good consequences. Receiving such a result would be like receiving a death sentence or a sentence of extended punishment. It will only engender feelings of hopelessness and depression and may cause some people to terminate their own lives prematurely.

*28. A 45-year-old woman undergoes genetic testing and discovers that she is at high risk for developing colon cancer and Alzheimer disease. Because her children have 50% of her genes, they also may be at an increased risk for these diseases. Does she have a moral or legal obligation to tell her children and other close relatives about the results of her genetic testing?

Solution:

Legally, she is not required to inform her children or other relatives about her test results, but people may have different opinions about her moral and parental responsibilities. On the one hand, she has the legal right to keep private the results of any medical information, including the results of genetic testing. On the other hand, her children may be at an increased risk of developing these disorders and might benefit from that knowledge. For example, the risk of colon cancer can be reduced by regular examinations so that tumors can be detected and removed before they become cancerous. Some people might argue that her parental responsibilities include providing her children with information about possible medical problems. Another issue to consider is the possibility that her children or other relatives might not want to know their genetic risk, particularly for a disorder such as Alzheimer disease for which there is no cure.

*29. Suppose that you could undergo genetic testing at age 18 for susceptibility to a genetic disease that would not appear until middle age and has no available treatment.

**a.** What would be some of the possible reasons for having or not having such a genetic test?

Solution:
Having the genetic test removes doubt about the potential for the disorder: you are either susceptible or not. Knowing about the potential of a genetic disorder enables you to make lifestyle changes that might lessen the effect of the disease or lessen the risk. The types and nature of future medical tests could be positively affected by the genetic testing, thus allowing for early warning and screening for the disease. The knowledge could also enable you to make informed decisions about whether to have children and the potential of passing the trait to your offspring. Additionally, by knowing what to expect, you could plan your life accordingly

Reasons for not having the test typically concern the potential for testing positive for the susceptibility to the genetic disease. If the susceptibility is detected, there is potential for discrimination. For example, your employer (or possibly a future employer) might consider you a long-term liability, thus affecting employment options. Insurance companies may not want to insure you for the condition or its symptoms, and social stigmatism regarding the disease could be a factor. Knowledge of the potential future condition could lead to psychological difficulties in coping with the anxiety of waiting for the disease to manifest.

**b.** Would you personally want to be tested? Explain your reasoning.

Solution:
There is no "correct" answer, but some of the reasons for wanting to be tested are: The test would remove doubt about the susceptibility, particularly if family members have had the genetic disease; either a positive or negative result would allow for informed planning of lifestyle, medical testing, and family choices in the future.