Chapter 02: Suggested Answers to In-Text Questions Biological and Environmental Foundations

Table of Contents

Ethical and Policy Applications of Lifespan Development: Prenatal Sex Selection – What Do You	J
Think? (p. 40)	. 1
Thinking in Context 2.1 (p. 41)	. 1
Thinking in Context 2.2 (p. 43)	. 2
Thinking in Context 2.3 (p. 50)	. 2
Lives in Context: Gene–Environment Interactions and Responses to Child Maltreatment – Wha	it
Do You Think? (p. 52)	. 3
Applying Developmental Science: Altering the Epigenome - What Do You Think? (p. 54)	. 4
Thinking in Context 2.4 (p. 56)	. 4
Apply Your Knowledge (p. 56)	

Ethical and Policy Applications of Lifespan Development: Prenatal Sex Selection – What Do You Think? (p. 40)

- 1. What do you think about parents choosing the sex of their children? In your view, under what conditions is sex selection acceptable?
 - Student answers will vary. Look to see if the student takes one stance and then gives several examples to back up their reason.
- 2. If you were able to selectively reproduce other characteristics, apart from sex, what might you choose? Why or why not?
 - Student answers will vary. Look to see if the student has several reasons to back up their ideas.

Thinking in Context 2.1 (p. 41)

- 1. What is the difference between mitosis and meiosis? What purpose might each serve in transmitting genes? Why do you think they evolved?
 - Mitosis occurs when a DNA molecule divides and replicates itself. In meiosis, the DNA begins to divide and replicate itself but it pairs with the DNA of another cell that is also dividing. The two pairs come together to form a new, unique cell. In mitosis, division of DNA results in the same DNA, but in meiosis, the division results in a unique DNA that is the combination of two different DNA. In meiosis, two different DNA combines, so if one's DNA was abnormal, the new DNA may counter the

abnormal part of the other. However, if both have an abnormality that pairs together, it may be more likely to be expressed in meiosis. These probably evolved to overcome some abnormalities by having one part of the DNA counter the abnormal part of the other.

- 2. In your view, why does twinning occur? From an evolutionary developmental perspective, does twinning serve an adaptive purpose for our species? Why or why not?
 - According to the text, twinning is more likely to occur in women over the age of 35. In this case, it may serve as an adaptive purpose for our species. If a woman does not have children before the age of 35, then she can probably have more babies in order to continue to populate the world with the human species.

Thinking in Context 2.2 (p. 43)

- 1. Consider your own physical characteristics, such as hair and eye color. Are they indicative of recessive traits, or dominant ones?
 - The answer to this question will vary from student to student. If a student has brown or black hair or eyes, then those are dominant. If a student has blonde or red hair or blue or green eyes, then those are recessive.
- 2. Do you think that you might be a carrier of recessive traits? Why or why not?
 - The answer to this question will vary from student to student depending on the genetic make-up of the family.

Thinking in Context 2.3 (p. 50)

- 1. Discuss how PKU illustrates the following two themes in human development: (1) the role of nature and nurture in development and (2) interactions among domains of development.
 - A person with PKU will have many physical and health problems if the person does not get put on the right diet. The right diet decreases the chances of the person having severe difficulties.
- 2. Identify risk factors for genetic and chromosomal disorders. What can prospective parents do to minimize the risks? What specific advice do you give?

- The risk factors depend on whether or not a woman is over the age of 35 or if disorders run in one side or both sides of the family. Prospective parents can have tests done to determine whether or not the baby has a disorder. In many cases, treatments may be available for the baby during the fetal period. In other cases, therapy and treatment is provided once the baby is born.
- 3. Suppose you are a 36-year-old woman pregnant with your first child. Considering the four types of prenatal diagnostic testing described in this section, what would be the advantages and disadvantages of each? What information would your health care provider need in order to recommend testing appropriate for your particular case?
 - Ultrasound: Advantage it can detect physical abnormalities. Disadvantage radiation can be harmful.
 - Amniocentesis: Advantage it can detect abnormalities that ultrasound may not. Disadvantage – higher risk of miscarriage.
 - Chorionic villus sampling: Advantage it can be done before amniocentesis. Disadvantage higher risk of damage to limbs or miscarriage.
 - Noninvasive prenatal testing (NIPT): This can be done simply by drawing the mother's blood after the 10th week of pregnancy. Disadvantage – it doesn't detect as many abnormalities as amniocentesis.
 - The healthcare provider would need to know the age of the mother and whether or not abnormalities run in the family of the mother or father.

Lives in Context: Gene–Environment Interactions and Responses to Child Maltreatment – What Do You Think? (p. 52)

- 1. In your view, how important are genetic contributors to development?
 - Genetic contributors are important. The reasoning will vary depending on students, but answers should include several reasons to back up their opinion.
- 2. If some genes may be protective in particular contexts, should scientists learn how to turn them on? Why or why not? What about genes that may be harmful in particular contexts?
 - Student opinions on this question will vary. Some may vary due to religious beliefs. The answers to this question should take into context

the students' personal beliefs and whether or not the rest of the answer supports the reasoning.

Applying Developmental Science: Altering the Epigenome - What Do You Think? (p. 54)

- 1. Much of the research on epigenetics examines animals, but there is a growing body of work studying humans. In what ways, if any, might you expect research findings based on people to differ from the findings of animal research, described previously? Explain.
 - Due to ethics, not all research done on animals can be done on humans.
 Additionally, it would not be ethical to put a human in a situation to determine what turns on or off certain genes.
- 2. What might you do to "care for" your epigenome? Identify activities and behaviors that you think might affect the health of your genome today and tomorrow.
 - One would have to know what issues run in his or her family and how they
 are influenced by the environment. Then the person would have to be
 aware of that in order to keep from having the environment influence his
 or her behavior.

Thinking in Context 2.4 (p. 56)

To answer the following questions, begin by thinking about how your own development reflects interactions among your genes and sociocultural context. Then, describe a skill, ability, or hobby in which you excel.

- 1. How might a passive genetic-environment correlation account for this ability? For example, in what ways has the context in which you were raised shaped this ability?
 - This answer will vary depending on students. Grading this answer should take into consideration whether or not the students' answers support their opinion.
- 2. In what ways might this ability be influenced by an evocative-geneticenvironment correlation?

- This answer will vary depending on students. Grading this answer should take into consideration whether or not the students' answers support their opinion.
- 3. Provide an example of how this ability might reflect an active geneticenvironment correlation.
 - This answer will vary depending on students. Grading this answer should take into consideration whether or not the students' answers support their opinion.
- 4. Which genetic-environment correlation do you think most accurately accounts for your skill, ability, or hobby?
 - This answer will vary depending on students. Grading this answer should take into consideration whether or not the students' answers support their opinion.
- 5. How might you apply the epigenetic framework to account for your ability?
 - This answer will vary depending on students. Grading this answer should take into consideration whether or not the students' answers support their opinion.

Apply Your Knowledge (p. 56)

- 1. Do you think Jenna and Tasha are MZ or DZ twins? Why or why not?
 - MZ twins. Although they do not look alike, many environmental factors contributed to that.
- 2. What role might epigenetic influences play in determining Jenna and Tasha's development?
 - Their environment influenced their development: extracurricular interests and skin tone for example. Nature influenced their intelligence.

Chapter 02: Web Exercises Biological and Environmental Foundations

Web Exercise #1: Sex Determination

LO 2.2. Identify and compare two processes of cell reproduction.

This chapter introduces students to two processes of cell reproduction: mitosis and meiosis. Further, it describes the importance of the 23rd pair of chromosomes, which determines sex of a baby. In some areas of the world, people use prenatal sex selection to ensure mothers give birth to male children. The textbook cites an essay (Bhatia, 2010) about China's one-child family policy, and information about the ethics of infanticide is located here: http://www.bbc.co.uk/ethics/abortion/medical/infanticide 1.shtml

In this assignment, students shall write a five-paragraph essay in support of or in argument against prenatal sex selection. Instructors may randomly select students to take one side or the other in order to debate the issue in class after students complete the assignments.

- 1. Search the reference section of the text to find the citation for Bhatia's article.
- 2. Search the university's library database to download a copy of Bhatia's article.
- 3. Read the information on infanticide provided in the link above.
- 4. Find additional resources to support or argue against prenatal sex selection.
- 5. Write a 500 750 word five-paragraph essay.

More information on writing five-paragraph essays may be found here: http://www.studygs.net/fiveparag.htm

Web Exercise #2: Switched at Birth

LO 2.3. Differentiate monozygotic (MZ) from dizygotic (DZ) twins.

The textbook discusses the differences between monozygotic (MZ) and dizygotic (DZ) twins. MZ twins are the product of a single fertilized egg (zygote) dividing and becoming two genetically identical individuals; yet, DZ twins are the product of two zygotes developing into individuals, who are not genetically identical. The purpose of this activity is to think about the lesson in this chapter and make connections to the lessons presented in Chapter 1.

Read the following *New York Times* article from July 2015: The Mixed up Brothers of Bogota

http://www.nytimes.com/2015/07/12/magazine/the-mixed-up-brothers-of-bogota.html? r=0

- 1. Compare and contrast the similarities and differences in each set of twins raised together as DZ twins.
- 2. Compare and contrast the similarities and differences of each set of MZ twins.
- 3. Relating this topic to Chapter 1, think about the nature vs. nurture debate and Bronfenbrenner's bioecological theory. How may similarities and differences in the brothers be explained by these theories?
- 4. Relating this topic to Chapter 1, how has the context of development in the lives of each man changed?
- 5. What do you think will be cognitive and social challenges each man will face?

Web Exercise #3: Genetic Disorders and Eugenics

LO 2.6. Identify disorders that result from chromosomal abnormalities.

LO 2.7. Discuss genetic counseling and prenatal testing, including common prenatal tests.

As noted in the chapter, some disorders are the result of chromosomal abnormalities. Several factors contribute to genetic disorders, and many prenatal tests now reveal abnormalities. Parents at risk of having children with genetic disorders or who have children with genetic disorders now have the availability to receive genetic counseling and therapy for their children. However, these options only recently became available. The United States has a history of supporting a eugenics program, which influenced Hitler's plot to create a superior human race. For more information, view this website:

http://historynewsnetwork.org/article/1796

- 1. Discuss the history of the United States eugenics program.
- 2. Who financed it?
- 3. How did it influence Hitler?
- 4. Do you think eugenics programs no longer exist? In what ways do they?
- 5. What are the pros and cons of the Human Genome Project?

Chapter 02: Chapter Exercises Biological and Environmental Foundations

Chapter Exercise #1: Nature vs. Nurture and Twins

LO 2.3: Differentiate monozygotic (MZ) from dizygotic (DZ) twins.

Directions: This assignment can be completed independently or in small groups. Chapter 1 introduces students to the study of nature versus nurture in human development. Several types of studies are performed to determine whether or not nature or nurture more strongly predicts human behavior or cognition. Create an idea for a research study using MZ and DZ twins to determine whether or not nature or nurture predicts performance of a behavior or cognitive ability.

- 1. Identify the target behavior or cognitive ability.
- 2. Develop a hypothesis.
- 3. How many MZ twins and how many DZ twins do your plan to study? Are they males or females? How old are they? Where do you plan to find them? Will they be paid to participate in this study? If so, how much?
- 4. Do you plan to use any other people? Any non-related participants? Any people who were adopted? Why or why not?
- 5. What tests or procedures will you use to determine whether or not nature or nurture more strongly predicted performance or ability?

Chapter Exercise #2: Genetic Family Tree

LO 2.4: Contrast four processes of genetic inheritance.

Directions: This assignment must be completed independently.

Materials: Microsoft PowerPoint.

Chapter 2 introduces students to the process of genetic inheritance. It discusses dominant and recessive genes, alleles, and how traits are expressed. Think about your own family or create a case. Find photos of family members or create a hypothetical family (students may use photos from Facebook or may Google images to construct a hypothetical family or to construct profiles of unknown biological relatives).

- 1. Describe each member's features: a) skin tone, b) eye color, c) hair color, and d) height. Discuss similarities in features.
- 2. What characteristics are dominant? What features are recessive?
- 3. Who in the family has a phenotype expressing recessive alleles? Why?
- 4. What characteristics illustrate polygenic inheritance? What patterns of inheritance are illustrated?

Chapter Exercise #3: Chromosomal Disorders

LO 2.6: Identify disorders that result from chromosomal abnormalities.

Directions: This assignment may be completed individually or in small groups.

Table 2.3 provides an overview of several diseases inherited through dominant-recessive inheritance. The purpose of this assignment is to develop a better understanding of one of these diseases. Students may answer the following questions or create a presentation on a three-sided poster board to present to the class.

- 1. Choose a disease from Table 2.3.
- 2. When was this disease discovered?
- 3. What is the mode of inheritance?
- 4. When may a person be diagnosed as having the disease? What are the criteria for diagnosis?
- 5. Give a brief description of the disease.
- 6. Is there a treatment? If so, what is it?
- 7. What is the life expectancy of a person with this disease?
- 8. What psychological or social challenges might someone with this disease face?
- 9. Who are some famous people with this disease?

Chapter 02: Discussion Questions Biological and Environmental Foundations

Discussion Question #1

This is a psychology/human development/education class. Why do you think it is important to include a chapter about biological and environmental foundations? What might these contribute to psychological development? What might these contribute to social development?

Discussion Question #2

Think about your own appearance. How similar are you to your siblings, your parents, your aunts and uncles, and your grandparents? What features are dominant? What features are recessive? If you could change one allele in your appearance, what would it be? Why? If you could change one allele in your family, what would it be? Why?

Discussion Question #3

Did you know that the United States once had an eugenics program in the early 1900s? Thousands of Americans were sterilized to ensure their genes were not passed down to another generation. Later, Hitler used eugenics to try to create a better human race. Do you feel it is ok to sterilize some humans? Why or why not?

Discussion Question #4

Do you know of anyone with a genetic disorder? Think about his/her psychological or social development. What challenges has this person faced? Why is it important to learn about these disorders in a class about lifespan development? What can a person with a genetic disorder teach us about life and the way "normal" people view it?

Discussion Question #5

This chapter discusses how heredity and the environment influence each other. In what ways does the environment influence heredity? In what ways does heredity influence the environment? In what ways have your environment influenced your genes? In what ways have your genes influenced your environment?

Discussion Question #6

Think about passive, evocative, and active gene-environment correlations. In what ways have your parents shaped your environment? How has your development been evocative? How have you actively shaped your environment? Can you identify personal examples of each gene-environment correlation?

Chapter 02: Discussion Questions Biological and Environmental Foundations

Learning Objectives

- **2.1.** Explain how chromosomes, genes, DNA, and the genome relate to one another.
- **2.2.** Identify and compare two processes of cell reproduction.
- **2.3.** Differentiate monozygotic (MZ) from dizygotic (DZ) twins.
- **2.4.** Contrast four processes of genetic inheritance.
- 2.5. Provide examples of diseases that illustrate dominant–recessive and X-linked inheritance.
- **2.6.** Identify disorders that result from chromosomal abnormalities.
- **2.7.** Discuss genetic counseling and prenatal testing, including common prenatal tests.
- **2.8.** Describe the methods and major findings of behavior genetics.
- **2.9.** Compare patterns of gene expression, including reaction range, canalization, gene–environment correlations, and epigenetic framework.

Chapter Summary

2.1. Explain how chromosomes, genes, deoxyribonucleic acid (DNA), and the genome relate to one another.

The human body is composed of cells. Within each cell is a nucleus that contains 23 matching pairs of rod-shaped structures called chromosomes. Each chromosome holds the basic units of heredity, known as genes. Genes are composed of stretches of DNA, a complex molecule shaped like a twisted ladder or staircase. Genes are the blueprint for creating all of the traits that organisms carry. The set of instructions to construct a living organism is referred to as the genome. Although all humans share the same basic genome, every person has a slightly different code, making him or her genetically distinct from other humans.

2.2. Identify and compare two processes of cell reproduction.

Most cells in the human body reproduce through mitosis in which DNA replicates itself, permitting the duplication of chromosomes, and ultimately the formation of new body cells with identical genetic material. Gametes reproduce by meiosis in which the chromosomes cross over as they replicate, creating two cells that each have 46 chromosomes; they then divide a second time, creating gametes with 23 single, unpaired chromosomes. Gametes have only one chromosome from each pair. Sperm and ovum join to produce a fertilized egg with 46 chromosomes, forming 23 pairs with half from the biological mother and half from the biological father.

2.3. Differentiate monozygotic (MZ) from dizygotic (DZ) twins.

Twins are siblings who share the same womb. Most are DZ twins who arise when a woman releases more than one ovum. Two ova are fertilized by two sperm. Genetically, DZ twins are no different from other siblings, sharing one half of their genes. Like other siblings, most differ in appearance, such as hair color, eye color, and height, but some are very similar. Unlike DZ twins, MZ twins originate from the same zygote. MZ twins arise when the zygote splits into two distinct separate yet identical zygotes that

give rise to two births. MZ twins share the same genotype with identical instructions for all physical and psychological characteristics. They share phenotypes that are carried by genes.

2.4. Contrast four processes of genetic inheritance.

Some genes are passed through dominant–recessive inheritance, in which some genes are dominant and will always be expressed regardless of the gene it is paired with. Other genes are recessive and will only be expressed if paired with another recessive gene. When a person is heterozygous for a particular trait, the dominant gene is expressed and the person remains a carrier of the recessive gene. In most cases, dominant–recessive inheritance is an oversimplified explanation for patterns of genetic inheritance. Incomplete dominance is a genetic inheritance pattern in which both genes influence the characteristic. In addition, most traits are a function of the interaction of many genes, known as polygenic inheritance. Researchers cannot trace most characteristics to only one or two genes. Instead, polygenic traits are the result of interactions among many genes. Some traits are determined by a process known as genomic imprinting. Genomic imprinting refers to the instance in which the expression of a gene is determined by whether it is inherited by the mother or the father.

2.5. Provide examples of diseases that illustrate dominant-recessive and X-linked inheritance.

PKU is a recessive disorder that occurs when both parents carry the allele. Individuals with PKU cannot produce an enzyme that breaks down phenylalanine. The phenylalanine builds up quickly to toxic levels that damage the central nervous system. Avoiding sources of phenylalanine can prevent some of the deficits. Disorders carried by dominant alleles, such as Huntington's disease on the other hand, are expressed when the individual has a single allele. Some recessive genetic disorders, like the gene for red-green color blindness, are carried on the X chromosome. Males are more likely to be affected by X-linked genetic disorders, such as hemophilia, because they have only one X chromosome. Females have two X chromosomes; a recessive gene located on one X chromosome will be masked by a dominant gene on the other X chromosome. Fragile X syndrome, on the other hand, is an example of a dominant-recessive disorder carried on the X chromosome. Because the gene is dominant, it must appear on only one X chromosome to be displayed. That means that fragile X syndrome occurs in both males and females

2.6. Identify disorders that result from chromosomal abnormalities

Klinefelter syndrome occurs in males born with an extra X chromosome (XXY). Symptoms range in severity, but most men show few symptoms that impair daily life and others may be unaware of the disorder until they are tested for infertility. A second type of sex chromosome abnormality experienced by men is XYY syndrome, also known as Jacob's syndrome, a condition that causes men to produce high levels of testosterone. Females are susceptible to a different set of sex chromosome abnormalities. Some females are born with three X chromosomes, known as triple X syndrome. Turner syndrome occurs when a female is born with only one X chromosome. The most common chromosome disorder is trisomy 21, known as Down syndrome. Down syndrome occurs when three chromosomes, rather than two, appear in place of the 21st pair of chromosomes. Although individuals with Down syndrome vary in the severity of symptoms, Down syndrome is associated with marked physical, health, and cognitive attributes.

2.7. Discuss genetic counseling and prenatal testing, including common prenatal tests.

Genetic counseling is a medical specialty that helps prospective parents determine the likelihood that their children will inherit genetic defects and chromosomal abnormalities. Prenatal testing is recommended when genetic counseling has determined a risk for genetic abnormalities or when fetal development appears abnormal. The most widespread and routine diagnostic procedure is ultrasound, in which high-frequency sound waves directed at the mother's abdomen provide clear images of the womb represented on a video monitor. Amniocentesis is a prenatal diagnostic procedure in which a small sample of the amniotic fluid that surrounds the fetus is extracted from the mother's uterus in order to conduct analyses to detect genetic and chromosomal anomalies and defects. CVS can be

conducted earlier than amniocentesis, between 9 and 12 weeks of pregnancy. CVS requires studying a small amount of tissue from the chorion, part of the membrane surrounding the fetus, for the presence of chromosomal abnormalities. Advances in genetics and in medicine have led to new therapies to reduce the effects of many genetic abnormalities, such as administering drugs to the fetus and conducting fetal surgery. One day we may be able to treat many heritable disorders through genetic engineering.

2.8. Describe the methods and major findings of behavior genetics.

Behavior genetics is the field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors. Even traits with a strong genetic component are modified by environmental influences. Heritability refers to the extent to which variation among people on a given characteristic is due to genetic differences. Heritability research examines the contributions of the genotype in determining phenotypes but also provides information on the role of experience. In selective breeding studies, behavior geneticists deliberately modify the genetic makeup of animals to examine the influence of heredity on attributes and behavior. Family studies, such as twin and adoption studies, compare people who live together and share varying degrees of relatedness. Twin designs compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes. Adoption studies compare the degree of similarity between adopted children and their biological parents whose genes they share and their adoptive parents with whom they share no genes. Genetics contributes to many traits, such as intellectual ability, sociability, anxiety, agreeableness, activity level, obesity, and susceptibility to various illnesses, yet even identical twins who share 100% of their genes are not 100% alike.

2.9. Compare patterns of gene expression, including reaction range, canalization, gene-environment correlations, and epigenetic framework.

Reaction range refers to the idea that there is a wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints. Some traits illustrate canalization and require extreme changes in the environment to alter their course. Gene—environment correlations illustrate how traits often are supported by both our genes and environment. Passive gene—environment correlations occur because parents are genetically similar to their children, the homes that they create in line with their own interests and preferences also correspond with and support the child's genotype. In a reactive or evocative gene—environment correlation, a child's genetic traits, such as personality characteristics, influence the social and physical environment, which shape development in ways that support the genetic trait. An active gene—environment correlation occurs when the child actively creates experiences and environments that correspond to and influence his or her genetic predisposition (called niche-picking). The epigenetic framework is a model for understanding the dynamic ongoing interactions between heredity and environment. Genes influence development and experience, yet gene expression is also influenced by development and experience.

Lecture Notes

- I. Genetic Foundations of Development
 - A. The Genetic Code:
 - a. Chromosomes
 - b. Genes
 - c. DNA
 - d. Genome
 - B. Cell Reproduction
 - a. Mitosis
 - b. Gametes

- c. Meiosis
- d. Crossing-over
- e. Zygote
- C. Sex Determination
 - a. The 23rd pair of chromosomes specify the biological sex of an individual
 - b. Sex is determined by whether or not the sperm carries an X or Y chromosome
 - i. XX = female
 - ii. XY = male
- D. Genes Shared by Twins
 - a. Dizygotic (DZ) twins
 - i. Fraternal twins
 - ii. Conceived when a woman releases more than one ovum and each is fertilized by a different sperm
 - iii. No more similar to each other than are other siblings that are conceived and born separately
 - b. Monozygotic (MZ) twins
 - i. Identical twins
 - ii. Originate from the same zygote, which splits into two distinct separate but identical zygotes that develop into two infants
 - iii. Share the same genotype with identical instructions for all physical and psychological characteristics
- II. Patterns of Genetic Inheritance
 - A. Dominant-Recessive Inheritance
 - a. Alleles
 - b. Homozygous alleles
 - c. Heterozygous alleles
 - d. Dominant-recessive inheritance
 - e. Carrier
 - B. Incomplete Dominance
 - a. A genetic inheritance pattern in which both genes influence the characteristic
 - b. One allele is stronger than the other yet does not completely dominate
 - c. Example: sickle cell alleles
 - C. Polygenetic Inheritance
 - a. Most traits are the function of the interaction of many genes
 - b. Examples: height, intelligence, temperament, and susceptibility to certain forms of cancer
 - D. Genomic Imprinting
 - a. The instance in which the expression of a gene is determined by whether it is inherited from the mother or the father
 - b. May influence susceptibility to illness such as some cancers
 - c. Examples: Prader-Willi syndrome & Angelman syndrome
- III. Chromosomal and Genetic Problems
 - A. Diagnosis
 - a. Many hereditary and chromosomal abnormalities can be diagnosed prenatally.
 - i. Routine blood tests
 - ii. Ultrasound examinations
 - iii. Specific tests recommended for couples who have a family history of such abnormalities or in which the woman is over the age of 35.

- b. Some hereditary and chromosomal abnormalities are evident at birth.
- c. Some hereditary and chromosomal abnormalities are detected soon after an infant begins to develop.
- d. Others reveal themselves only over a period of years
- B. Dominant-Recessive Disorders
 - a. Cystic fibrosis
 - b. Sickle cell anemia
 - c. Few severe disorders are inherited through dominant inheritance because individuals who inherit the allele often do not survive long enough to reproduce and pass it to the next generation. (Example: Huntington's disease)
 - d. Phenylketonuria (PKU)
 - a. Causes mental retardation
 - b. Example of how genes interact with the environment
 - i. Intellectual disability results from the interaction of the genetic predisposition and exposure to phenylalanine from the environment.
 - Placed on a diet low in phenylalanine and attain average or nearaverage levels of intelligence
 - c. Difficulty in attention and planning
 - d. Behavioral and psychological problems, including sadness, depression, anxiety, and phobic reactions
 - e. Emotional and social challenges due to strict diet
- C. X-Linked Disorders
 - a. Due to genes on X chromosome
 - b. Color blindness
 - c. Hemophilia
 - i. Blood does not clot normally
 - ii. Females have it only if both X chromosomes have the gene.
 - iii. Males have it because the Y chromosome cannot have genetic information to counter it.
 - d. Fragile X
 - i. Due to a pinched site toward the end of the X chromosome
 - ii. Dominant and needs only to appear on only one X chromosome to be displayed
 - iii. Most common form of inherited intellectual impairment
 - iv. May be associated with autism
 - v. Cardiac defects are common
 - vi. Poor eye contact and repetitive behaviors
- D. Chromosomal Abnormalities
 - a. Down syndrome
 - i. Occurs when three chromosomes, rather than two, appear in place of the 21st pair of chromosomes
 - ii. Show delays in physical and motor development relative to other children
 - iii. Experience immune system deficiencies and frequent infections, especially of the respiratory tract
 - iv. Intellectual disabilities most common genetic cause of mental retardation
 - v. Need parents who are sensitive to their capacities and encourage them
 - vi. Lifespan has increased from 25 years in 1980s to an expectancy of 60 today
 - vii. At risk to show signs of Alzheimer's very early relative to other adults

E. Sex chromosome Abnormalities

- a. Klinefelter syndrome
 - i. Males are born with an extra X chromosome.
 - ii. One of the most common sex chromosome abnormalities
 - iii. Feminine body shape, high pitched voice
 - iv. Infertile
- b. Jacob's syndrome
 - i. Male produces high levels of testosterone.
 - ii. Severe acne
 - iii. Poor coordination
- c. Triple X syndrome
 - i. Women appear normal
 - ii. Low range of intelligence
 - iii. Small learning difficulties
- d. Turner syndrome
 - i. Female has only one X chromosome
 - ii. Short
 - iii. Ovaries do not develop normally; do not ovulate
 - iv. Spontaneous onset of puberty instead of gradual
 - v. Regular injections of human growth hormones can result in some breast development and menstruation

F. Mutation

- b. Sudden changes and abnormalities of genes
- a. May involve one gene or many
- b. May occur spontaneously or as a result of exposure to environmental toxins
- c. Most are fatal developing organism often dies very soon after conception, often before the woman knows she is pregnant
- d. Sometimes mutations are beneficial (Example: sickle cell results in resistance to malarial infection)
- G. Predicting and Detecting Genetic Disorders
 - a. Heritability
 - b. Genetic counseling
 - i. Helps prospective parents determine the risk their children will inherit genetic defects and chromosomal abnormalities
 - ii. Valuable when one or both prospective parents have relatives with inborn disorders
 - c. Prenatal Diagnosis
 - Recommended when genetic counseling has determined a risk for genetic abnormalities, when the woman is older than 35k, when both parents are members of an ethnicity at risk for particular genetic disorders, or when fetal development appears abnormal
 - ii. Can detect many defects
 - iii. Amniocentesis
 - iv. Chronic Villus Sampling (CVS)
 - v. Noninvasive Prenatal Testing (NIPT)
 - d. Prenatal Treatment of Genetic Disorders
 - i. Hormones
 - ii. Medications

- iii. Blood transfusions
- iv. Fetal surgery
- IV. Heredity and Environment
 - a. Phenotypes are influenced by the interaction of genotypes and our experiences
 - A. Behavioral Genetics examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors
 - a. Methods of Behavior Genetics
 - i. Selective breeding
 - ii. Family studies
 - b. Genetic Influences on Personal Characteristics
 - i. Identical twins consistently have more highly correlated intellectual scores than do fraternal twins.
 - ii. Genes contribute to growth, body weight, and body height, but environmental influences also play a role.
 - B. Gene-Environment Interaction
 - a. Range of reaction
 - b. Canalization
 - c. Gene-environment correlations
 - i. Passive gene-environment
 - ii. Evocative gene-environment
 - iii. Active gene-environment
 - C. Epigenetic Framework
 - a. Development results from ongoing reciprocal interactions between genetics and environment
 - b. Not all genes are switched on at birth



DEVELOPMENT CONTEXT



Biological and Environmental Foundations

The Genetic Code

-Cells

 Each cell contains 23 matching pairs of chromosomes

Chromosomes

Holds the basic units of heredity (genes)

– Genes

Composed of stretches of DNA (deoxyribonuclei acid)

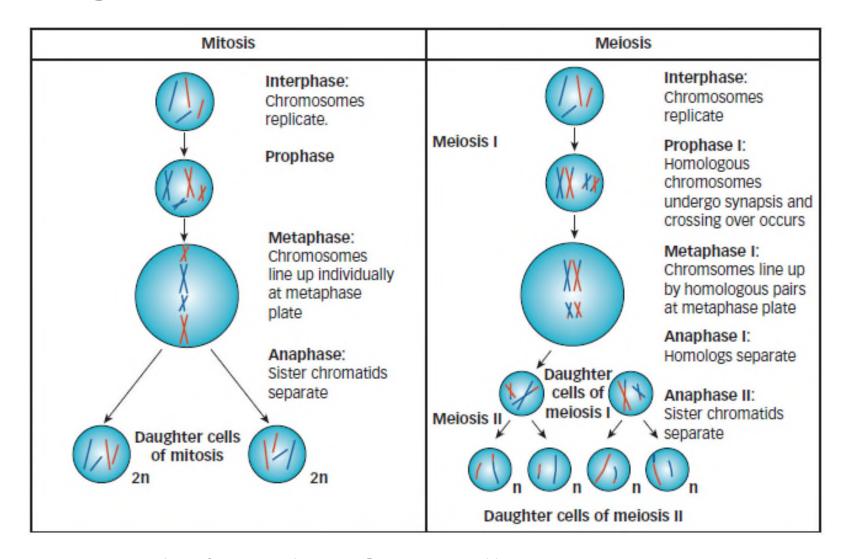
Genome

- A genome is the set of instructions to construct a living organism.
- Every species has a different genome.
- Humans share 99% of our DNA with the chimpanzee.
 - The 1% of our genes influence the characteristics that differentiate us from chimpanzees (i.e., cognitive and language abilities).

Cell Reproduction - Mitosis

- Most cells in the human body reproduce through mitosis.
- DNA replicates itself.

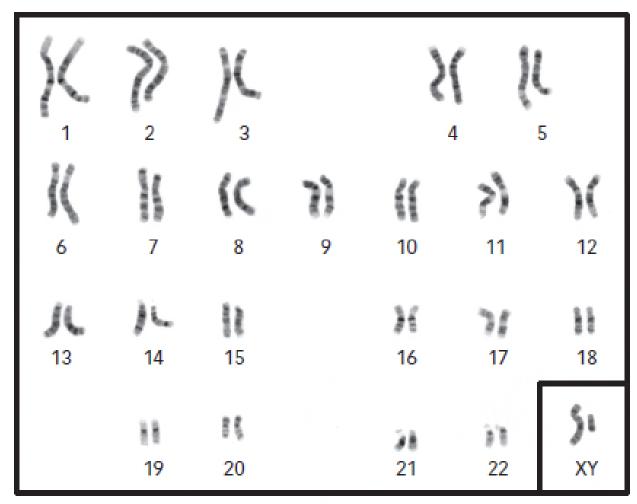
Figure 2.1: Mitosis and Meiosis



Cell Reproduction - Meiosis

- Sex cells (gametes sperm in males; ova in females) reproduce through meiosis
 - Gametes each contain 23 chromosomes.
 - Crossing-over creates unique combinations of genes (Chromosome pairs align, and DNA segments cross over, moving from one member of the pair to the other).
 - After crossing-over is complete, the cell then divides into two cells, each with 46 chromosomes.
 - As the new cells replicate, they create cells containing only 23 single, unpaired chromosomes.

Figure 2.2: Chromosomes



autosomes

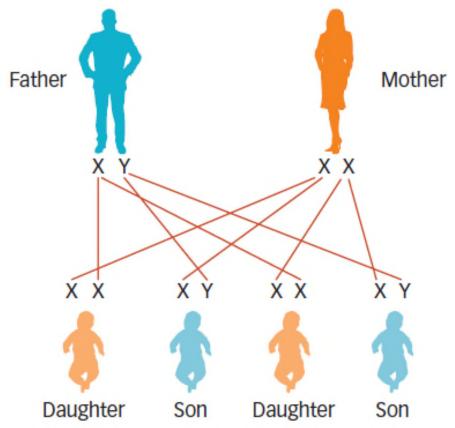
sex chromosomes

Zygote

- The fertilized egg
 - Contains 46 chromosomes, forming 23 pairs with half from the biological mother and half from the biological father

Figure 2.3: Sex Determination

• The sex chromosomes determine whether a zygote will develop into a male or female.



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Sex Determination (Continued)

- All ova contain one X sex chromosome.
- One half of the sperm males produce contain an X chromosome; one half contain a Y.
 - If the ovum is fertilized by a Y sperm, a male fetu will develop.
 - If the ovum is fertilized by an X sperm, a female fetus will form.

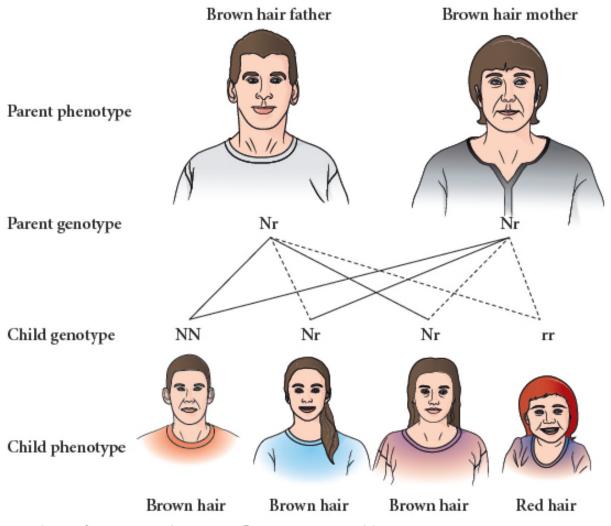
VIDEO CASE

Twins



Twins share both genes and environment. Velma and Thelma discuss their similarities and differences. Can you guess whether they are fraternal or identical twins?

Figure 2.4: Dominant-Recessive Inheritan



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Genes Shared by Twins

- Siblings who share the same womb
 - 1 out of every 30 births in the U.S.
- Dizygotic (DZ) twins
 - 13 of every 1,000 births
- Monozygotic (MZ) twins
 - 4 of every 1,000 births

Dizygotic (DZ) Twins

- Also known as fraternal twins
- Occur when more than one ovum is released and each is fertilized by a different sperm
- Genetically, DZ twins are no more similar to each other than siblings born separately
- DZ twins tend to run in families
- Rates of DZ twins increase with in vitro fertilization, maternal age, and with each subsequent birth

Monozygotic (MZ) Twins

- Also known as identical twins
- Twins originate from the same zygote
 - The zygote splits into two distinct separate but identical zygotes that develop into two infants
- MZ twins share the same genotype, with identical instructions for all physical and psychological characteristics
- The causes of not well understood, but chances increase with in vitro fertilization and increasing maternal age

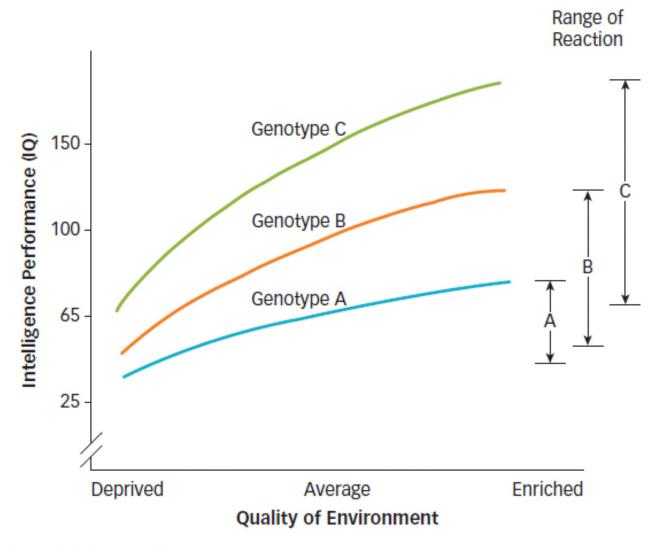
Dominant-Recessive Inheritance: Allele

- Genes expressed in different forms
- Influences a variety of physical characteristics
 - Homozygous alleles of the pair of chromosome are alike (e.g., hair color)
 - The person will display the inherited trait.
 - Heterozygous alleles of the pair of chromosome are different
 - The trait expressed will depend on the relations amor the genes.

Dominant-Recessive Inheritance: Dominant and Recessive Genes

- Dominant genes are always expressed regardless of the gene they are paired with.
- Recessive genes are only expressed if paired with another recessive gene.
- When an individual is heterozygous for a particular trait, the dominant gene is expressed and the person becomes a carrier of the recessive gene.

Figure 2.5: Illustration of Dominant-Recessive Inherita



SOURCE: Gottlieb (2007).

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Table 2.1: Dominant and Recessive Characteris

DOMINANT TRAIT	RECESSIVE TRAIT
Dark hair	Blond hair
Curly hair	Straight hair
Hair	Baldness
Non-red hair	Red hair
Facial dimples	No dimples
Brown eyes	Blue, green, hazel eyes
Second toe longer than big toe	Big toe longer than second toe
Type A blood	Type O blood
Type B blood	Type O blood
Rh-positive blood	Rh-negative blood
Normal color vision	Color blindness

SOURCES: McKusick (1998); McKusick-Nathans Institute of Genetic Medicine (2014).

Incomplete Dominance

- A genetic inheritance pattern in which both genes influence the characteristic
 - For example: blood type
- A different inheritance pattern is seen when a person inherits heterozygous alleles in which one allele is stronger than the other yet does not completely dominate.
 - For example: sickle cell anemia

Polygenic Inheritance

- Most traits are a function of the inheritance of many genes
- Examples include:
 - Height
 - Intelligence
 - Temperament
 - Susceptibility to certain forms of cancer

Genomic Imprinting

- The expression of a gene is determined by whether it is inherited from the mother or th father
- Genomic imprinting may influence susceptibility to illnesses

Table 2.3: Genetic Disorders: Dominant-Recessive Disorders

DISEASE	OCCURRENCE	MODE OF INHERITANCE	DESCRIPTION	TREATMENT
Huntington's disease	1 in 20,000	Dominant	Degenerative brain disorder that affects muscular coordination and cognition	No cure; death usually occurs 10 to 20 years after onset
Marfan syndrome	1 in 20,000	Dominant	A connective tissue disorder that affects the skeleton, lungs, eyes, heart and blood vessels; disease is characterized by unusually long limbs	No cure; death from complications in young adulthood is common
Cystic fibrosis	1 in 2,000-2,500	Recessive	An abnormally thick, sticky mucus clogs the lungs and digestive system, leading to respiratory infections and digestive difficulty	Bronchial drainage, diet, gene replacement therapy
Phenylketonuria (PKU)	1 in 8,000-10,000	Recessive	Inability to digest phenylalanine that, if untreated, results in neurological damage and death	Diet
Sickle cell anemia	1 in 500 African Americans	Recessive	Sickling of red blood cells leads to inefficient distribution of oxygen throughout the body that leads to organ damage and respiratory infections	No cure; blood transfusions, treat infections, bone marrow transplant; death by middle age
Tay-Sachs disease	1 in 3,600-4,000 descendants of Central and Eastern European Jews	Recessive	Degenerative brain disease	None; most die by 4 years of age
Cooley's anemia	1 in 500 people of Mediterranean descent	Recessive	Blood disorder resulting in very pale skin, retarded growth, and lethargy	No cure; frequent blood transfusions; death by complications occurs by adolescence

SOURCES: McKusick (1998); McKusick-Nathans Institute of Genetic Medicine (2014).

Table 2.4: Genetic Disorders: X-Linked Disord

SYNDROME/ DISEASE	OCCURRENCE	DESCRIPTION	TREATMENT
Color blindness	1 in 12 males	Difficulty distinguishing red from green; less common is difficulty distinguishing blue from green	No cure
Duchenne muscular dystrophy	1 in 3,500 males	Weakness and wasting of limb and trunk muscles; progresses slowly but will affect all voluntary muscles	Physical therapy, body braces; sur beyond late 20s
Fragile X syndrome	1 in 2,000 males	Symptoms include cognitive impairment; attention problems; anxiety; unstable mood; long face; large ears; flat feet; and hyperextensible joints, especially fingers	No cure
Hemophilia	1 in 3,000- 7,000 males	Blood disorder in which the blood does not clot	Blood transfusion

SOURCES: McKusick (1998); McKusick-Nathans Institute of Genetic Medicine (2014).

Chromosomal Abnormalities

- Occur when cells have too few or too many copies of a chromosome or when chromosomes are damaged or altered
 - Down Syndrome (1 of every 700 births)
 - Also known as trisomy 21 (three chromosomes appear in place of the 21st pair of chromosomes)
 - Risk increases with maternal age

Table 2.5: Sex Chromosome Abnormalit

FEMALE GENOTYPE	SYNDROME	MALE GENOTYPE	SYNDROME
XX	normal	XY	normal
XO	Turner	XXY	Klinefelter
XXX	Triple-X	XYY	XYY

Sex Chromosome Abnormalities - Male

- Klinefelter syndrome (XXY)
 - Most common sex chromosome abnormality
- Jacob's syndrome (XYY)
 - A condition that causes men to produce high levels of testosterone

Sex Chromosome Abnormalities - Fema

- —Triple X syndrome (XXX)
 - 1 in 1,000 females
 - Appear within the norm of other females
- Turner syndrome (0X)
 - Abnormal growth patterns

Mutation

- Sudden changes and abnormalities in the structure of genes
- May involve only one gene or many
- May occur spontaneously or by exposure to environmental toxins
- Common one half of all conceptions include mutated chromosomes (most are fatal)

Predicting and Detecting Genetic Disorders: Genetic Counseling

 A medical specialty that helps prospective parents determine the risk that their children will inherit genetic defects and chromosomal abnormalities

VIDEO CASE

Genetics and Pregnancy



Miscarriages occur for many reasons. In this video, Melody and Mark explain how they learned that their multiple miscarriages were the result of a genetic disorder.

Table 2.6: Predicting and Detecting Genetic Disorders: Prenatal Diagnosis

	EXPLANATION	ADVANTAGES	DISADVANTAGES	
Ultrasound	High-frequency sound waves directed at the mother's abdomen provide clear images of the womb projected on to a video monitor.	Ultrasound enables physicians to observe the fetus, measure fetal growth, reveal the sex of the fetus, and determine physical abnormalities in the fetus.	Many abnormalities and deformities cannot be easily observed.	
Amniocentesis	A small sample of the amniotic fluid that surrounds the fetus is extracted from the mother's uterus through a long, hollow needle inserted into the mother's abdomen. The amniotic fluid contains fetal cells. The fetal cells are grown in a laboratory dish in order to create enough cells for genetic analysis.	It permits a thorough analysis of the fetus's genotype. There is 100% diagnostic success rate.	It poses a greater risk to the fetus than ultrasound. If conducted before the 15th week of pregnancy, it may increase the risk of miscarriage.	
Chorionic villus sampling (CVS)	CVS requires studying a small amount of tissue from the chorion, part of the membrane surrounding the fetus, for the presence of chromosomal abnormalities. The tissue sample is obtained through a long needle inserted either abdominally or vaginally, depending on the location of the fetus.	It permits a thorough analysis of the fetus's genotype. CVS is relatively painless, and there is a 100% diagnostic success rate.	It may pose a higher rate of spontaneous abortion and limb defects when conducted prior to 10 weeks' gestation.	
Noninvasive prenatal testing (NIPT)	Cell-free fetal DNA are examined by drawing blood from the mother.	There is no risk to the fetus. It can diagnose several chromosomal abnormalities.	It cannot detect the full range of abnormalities. It may be less accurate than other methods.	

Behavior Genetics

- The field of study that examines how genes and experience combine to influence the diversity of human traits, abilities, and behaviors
 - Genotype
 - Genetic inheritance
 - Phenotype
 - The expression of genetic traits in observable characteristics and behaviors

Studies to Determine Relatedness

Family studies

- Twin studies
 - Compare identical and fraternal twins to estimate how much of a trait or behavior is attributable to genes
- Adoption studies
 - Compare the degree of similarity between adopted children and their biologcial parents whose genes the share (50%) and their adoptive parents with whom the share no genes

Table 2.7: Correlation of Intelligence Scores from Family Studies

	REARED TOGETHER	REARED APART
MZ twins (100% shared genes)	.86	.72
DZ twins (50% shared genes)	.60	.52
Siblings (50% shared genes)	.47	.24
Biological parent/child (50% shared genes)	.42	.22
Half-siblings (25% shared genes)	.31	-
Unrelated (adopted) siblings (0% shared genes)*	.34	_
Nonbiological parent/child (0% shared genes)*	.19	_

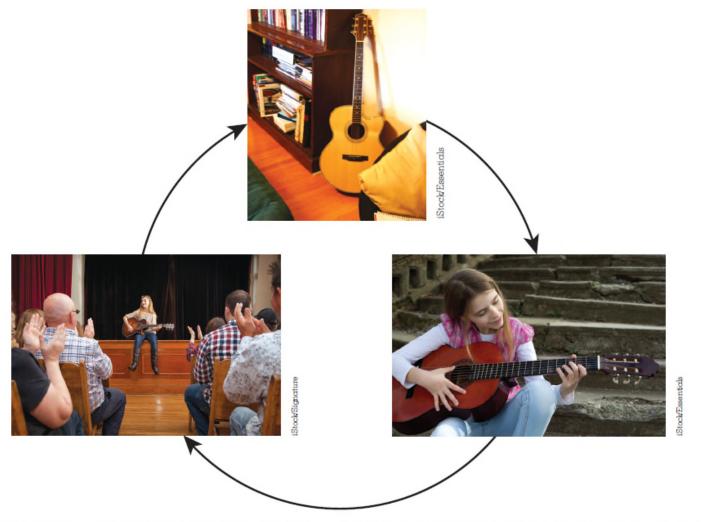
NOTES: *Estimated correlation for individuals sharing neither genes nor environment = .0; MZ = monozygotic; DZ = dizygotic.

SOURCE: Adapted from Bouchard and McGue (1981).

Gene-Environment Interaction: Range of Reaction

 A wide range of potential expressions of a genetic trait, depending on environmental opportunities and constraints

Figure 2.6: Gene-Environment Correlati



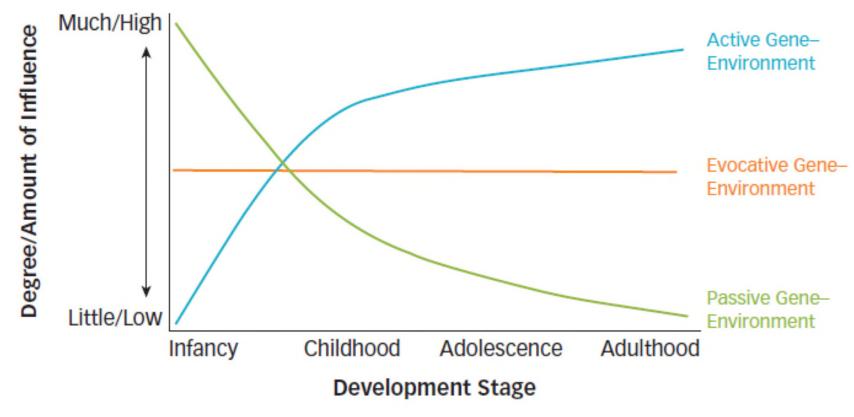
The availability of instruments in the home corresponds to the child's musical abilities and she begins to play guitar (passive gene-environment correlation). As she plays guitar, she evokes positive responses in others, increasing her interest in music (evocative gene-environment correlation). Over time she seeks opportunities to play, such as performing in front of an audience (niche picking).

Gene-Environment Interaction: Canalizati

- Heredity narrows the range of development to only one or a few outcomes
- Canalized traits are biologically programmed; only powerful environmental forces can change their developmental path

Gene-Environment Correlation

- The idea that many of our traits are supported by both our genes and environment
- Three types passive, reactive, and active



Epigenetic Framework

- The dynamic interplay between heredity and environment
- Environmental factors such as toxins, injuries crowding, diet, and responsive parenting can influence the expression of genetic traits
- Evocative gene-environmental correlations and niche-picking demonstrate ways genetic propensities can influence the environment