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### **Conception and Infertility**

*Learning Objective 2.1: Summarize how conception occurs and describe alternative paths to parenthood.*

- How does fertilization take place?
- What are some of the causes of infertility?
- How is infertility treated?
- What are alternative paths to parenthood?

### **Mechanisms of Heredity**

*Learning Objective 2.2: Explain how traits are passed down across generations.*

- How are genes inherited?
- What determines sexual differentiation?
- How are traits transmitted?

### **Genetic and Chromosomal Abnormalities**

*Learning Objective 2.3: Describe how abnormalities are transmitted in the genes and the options prospective parents have for testing for them.*

- What is dominant and recessive inheritance?
- What are sex-linked genetic defects?
- What are some common genetic abnormalities?
- How do we test for genetic abnormalities?

### **Studying the Influence of Heredity and Environment**

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*Learning Objective 2.4: Describe how researchers determine the relative influence of genes and environments, and how these variables interact with each other.*

- How do we measure the relative influences of genes and environment?

- How do genes and environments interact?

## Characteristics Influenced by Heredity and Environment

*Learning Objective 2.5: Summarize how genes affect physical, intellectual and personality development, as well as psychopathologies.*

- What individual characteristics are influenced by heredity/environment interactions?

## TOTAL TEACHING PACKAGE OUTLINE

### Chapter 2: Conception, Heredity, and Environment

<b>Learning Objective 2.1</b> Summarize how conception occurs and describe alternative paths to parenthood	<b>Discussion Topic 2.1 and 2.2</b> <b>Knowledge Construction Activity 2.2, 2.3, and 2.4</b>
<b>Learning Objective 2.2</b> Explain how traits are passed down across generations	<b>Knowledge Construction Activity 2.1</b>
<b>Learning Objective 2.3</b> Describe how abnormalities are transmitted in the genes and the options prospective parents have for testing for them	<b>Lecture Topic 2.1 and 2.2</b> <b>Discussion Topic 2.3</b> <b>Independent Study 2.1</b> <b>Knowledge Construction Activity 2.1 and 2.5</b>
<b>Learning Objective 2.4</b> Describe how researchers determine the relative influence of genes and environments, and how these variables interact with each other	<b>Knowledge Construction Activity 2.1 and 2.6</b>
<b>Learning Objective 2.5</b> Summarize how genes affect physical, intellectual and personality development, as well as psychopathologies	<b>Discussion Topic 2.4</b>
<b>Applied Activities</b>	<b>Applied Activity 2.1 and 2.2</b>

## EXPANDED OUTLINE

### I. Conception and Infertility

#### A. Fertilization

- **Fertilization**, or conception, is the process by which sperm and ovum—the male and female gametes, or sex cells—combine to create a single cell called a **zygote**, which then duplicates itself again and again by cell division to produce all the cells that make up a baby.
- At birth, a female is believed to have about 2 million immature ova in her two ovaries, each ovum in its own small sac, or follicle.
- In a sexually mature woman, ovulation occurs about once every 28 days until menopause.
- After being expelled from the ovary, the ovum is swept along through one of the fallopian tubes by tiny hair cells, called cilia, toward the uterus, or womb.
- Sperm are produced in the testicles (testes), or reproductive glands, of a mature male at a rate of several hundred million a day and are ejaculated in the semen at sexual climax.
  - Deposited in the vagina, they try to swim through the cervix (the opening of the uterus) and into the fallopian tubes.
- Fertilization typically occurs while the ovum is passing through the fallopian tube.
- If fertilization does not occur, the sperm are absorbed by the woman's white blood cells and the ovum passes through the uterus and exits through the vagina.

## B. Infertility

- The most common cause of infertility in men is a low sperm count or insufficiently motile (capable of motion) sperm.
  - A sperm count lower than 60 to 200 million per ejaculation makes conception unlikely.
- In a woman, common causes of infertility include:
  - The failure to produce eggs, or ova, or to the failure to produce normal ova
  - Mucus in the cervix, which prevents sperm from penetrating it
  - A disease of the uterine lining, which prevents implantation of the fertilized ovum
- A major cause of declining fertility in women after age 30 is deterioration in the quality of ova.
- However, the most common cause is blockage of the fallopian tubes, which prevents ova from reaching the uterus.

## C. Assisted Reproductive Technologies

- **Assisted reproductive technology (ART)**, or conception through artificial means, provides couples having difficulty conceiving naturally with a means to augment their

fertility.

- The simplest form of ART is *artificial insemination* in which sperm is injected into a woman's vagina, cervix, or uterus.
  - This procedure can facilitate conception if a man has a low sperm count.
- In another common method, *in vitro fertilization* (IVF), a woman first receives fertility drugs to stimulate the production of multiple ova.
  - Then the ova are surgically removed, fertilized in a laboratory dish, and implanted in the woman's uterus.
- IVF also addresses severe male infertility.
  - A single sperm can be injected into the ovum—a technique called intracytoplasmic sperm injection (ICSI).
- A woman who is producing poor-quality ova or who has had her ovaries removed may try ovum transfer.
  - In this procedure, a donor egg from a fertile younger woman is fertilized in the laboratory and implanted in the prospective mother's uterus.
  - Alternatively, the ovum can be fertilized in the donor's body by artificial insemination. The embryo is retrieved from the donor and inserted into the recipient's uterus.
- ART can result in a tangled web of legal, ethical, and psychological dilemmas.
  - The issues multiply when a *surrogate mother* is involved.
  - The surrogate, a fertile woman, is impregnated by the prospective father, usually by artificial insemination.
  - She agrees to carry the baby to term and give it to the father and his partner.

#### D. Adoption

- If a woman cannot conceive on her own, and she is either unwilling or unable to conceive with the ART, adoption is an alternative option.
- In the United States, adoptions may either be national or international.

## II. Mechanisms of Heredity

The science of genetics is the study of heredity—the inborn factors from the biological parents that affect development.

### A. The Genetic Code

- The stuff of heredity is a chemical called **deoxyribonucleic acid (DNA)**.

- The double-helix structure of DNA resembles a long, spiraling ladder whose steps are made of pairs of chemical units called *bases* (Figure 2.1).
- **Chromosomes** are coils of DNA that consist of smaller segments called **genes** and are found in every cell in the human body.
- Each gene has a specific location on its chromosome and contains thousands of bases.
- The complete sequence of genes in the human body constitutes the **human genome**.
- Every cell in the normal human body except the sex cells (sperm and ova) has 23 pairs of chromosomes—46 chromosomes in all.
- Through a type of cell division called *meiosis*, each sex cell ends up with only 23 chromosomes.
  - Thus, when sperm and ovum fuse at conception, they produce a zygote with 46 chromosomes: 23 from the father and 23 from the mother (Figure 2.2).
- Through a process known as *mitosis*, the DNA replicates itself so each newly formed cell is a complete genetic copy with the same hereditary information.

## B. Sex Determination

- Twenty-two of the 23 pairs of chromosomes are **autosomes**, chromosomes that are not related to sexual expression.
- The 23rd pair are **sex chromosomes**—1 from the father and 1 from the mother—that govern the baby's sex.
- Females have two X chromosomes (XX), and males have one of each type (XY).
- Each sperm cell has an equal chance of carrying an X or a Y, and thus it is the father who determines sex.
- Initially, the embryo's rudimentary reproductive system, which is basically female, appears almost identical in both males and females.
- Males' development requires the activation of the SRY gene.
  - Otherwise, male sexual development will not occur, and the embryo will develop genitals that appear female.
  - In normal development, male embryos start producing the hormone testosterone at about six to eight weeks after conception, resulting in the development of a male body with male sexual organs.
- The development of the female reproductive system is equally complex and depends on a number of genetic variants, including the *HOX genes* and a variety of signaling substances known as *Wnts*.

## C. Patterns of Genetic Transmission

- During the 1860s, Gregor Mendel, an Austrian monk, laid the foundation for the understanding of patterns of inheritance.
- By crossbreeding strains of peas, he discovered two fundamental principles of genetics:
  - Traits could be either dominant or recessive. *Dominant traits* are always expressed, while *recessive traits* are expressed only if both copies of the gene are recessive.
  - Traits are passed down independently of each other.

### 1. Dominant and Recessive Inheritance

- Genes that can produce alternative expressions of a characteristic, such as the presence or absence of dimples, are called **alleles**.
- Alleles are the different version of a particular gene.
- Every person receives one maternal and one paternal allele for any given trait.
- When both alleles are the same, the person is **homozygous** for the characteristic; when they are different, the person is **heterozygous**.
- In **dominant inheritance**, when an offspring receives at least one dominant allele for a trait, it will be expressed.
- **Recessive inheritance**, or the expression of a recessive trait, occurs only when a person receives two recessive alleles, one from each parent.
- Traits may also be affected by **mutations**, permanent alterations in genetic material.
  - Mutations, such as the spontaneous mutation known as achondroplasia which results in dwarfism, are generally due to copying errors and are usually harmful.

### 2. Multifactorial Transmission

- Most traits result from **polygenic inheritance**, the interaction of many genes.
  - For example, skin color is the result of three or more sets of genes on three different chromosomes.
  - These genes work together to produce different amounts of brown pigment, resulting in hundreds of shades of skin color.
  - This phenomenon is known as **multifactorial transmission**.

## D. Epigenesis: Environmental Influence on Gene Expression

- Your **genotype** is what is coded in your genes—the recipe for making you.
- What is expressed—who you actually are—is your **phenotype**.
- Except for monozygotic twins, identical twins who started out as a single fertilized ovum, no two people have the same genotype.



- The phenotype is the genotype in action.
- The difference between genotype and phenotype helps explain why a clone, a genetic copy of an individual, or even an identical twin can never be an exact duplicate of another person.
- Mounting evidence suggests that gene expression is controlled by reversible chemical reactions that turn genes on or off as they are needed but that do not change the underlying genetic code.
  - This phenomenon is called **epigenesis**.
- Epigenesis works via chemical molecules, or “tags,” attached to a gene that affect the way a cell “reads” the gene’s DNA.
- Because every cell in the body inherits the same DNA sequence, the function of the chemical tags is to differentiate various types of body cells.
- These tags work by switching particular genes on or off during embryonic formation.
- Epigenetic changes can occur throughout life in response to environmental factors such as nutrition, sleep habits, stress, and physical affection.
  - Sometimes errors arise, which may lead to birth defects or disease.
  - Epigenetic changes may also contribute to such common ailments as cancer, diabetes, and heart disease.
  - In addition, they may explain why one monozygotic twin is susceptible to a disease such as alcoholism but the other twin is not, and why some twins get the same disease but at different ages.
- Cells are especially susceptible to epigenetic modification during critical periods such as puberty and pregnancy.
  - Epigenetic changes may be heritable.

### III. Genetic and Chromosomal Abnormalities

- Soft markers are physical abnormalities that can be seen on an ultrasound; they indicate an increased risk of having a baby with a genetic disorder.
- Most birth disorders are fairly rare, affecting only about 3 percent of live births. Nevertheless, they are the leading cause of infant death in the United States, accounting in 2005 for 19.5 percent of all deaths in the first year in 2005.
- Not all genetic or chromosomal abnormalities are apparent at birth.
- Table 2.1 lists some of the disorders caused by genetic and chromosomal abnormalities.
- It is in genetic defects and diseases that we see most clearly the operation of dominant and recessive transmission, and also of a variation, sex-linked inheritance.

#### A. Dominant or Recessive Inheritance of Defects

- Most of the time, normal genes are dominant over those carrying abnormal traits, but sometimes the gene for an abnormal trait is dominant.
  - When this is the case, even one copy of the “bad” gene will result in a child expressing the disorder.
- Among the 1,800 disorders known to be transmitted by dominant inheritance are achondroplasia (a type of dwarfism) and Huntington’s disease.
- Although they can be serious, defects transmitted by dominant inheritance are less likely to be lethal at an early age than those transmitted by recessive inheritance.
  - This is because if a dominant gene is lethal at an early age, then affected children would be likely to die before reproducing.
- Recessive defects are expressed only if the child is homozygous for that gene; in other words, a child must inherit a copy of the recessive gene from each parent to be affected.
  - Because recessive genes are not expressed if the parent is heterozygous for that trait, both parents may be carriers without realizing it.
  - In this case, any child they had would have a 25 percent chance of getting both of the recessive copies, and thus expressing the trait.
  - Defects transmitted by recessive genes tend to be lethal at an earlier age, in contrast to those transmitted by dominant genes as they can be passed down to the next generation by carriers.
- In **incomplete dominance**, a trait is not fully expressed.

## B. Sex-Linked Inheritance of Defects

- Certain recessive disorders are transmitted by **sex-linked inheritance**.
  - They are linked to genes on the sex chromosomes and affect male and female children differently.
- When a mother is a carrier of a sex-linked disorder, she has a 50 percent chance of passing that gene on to her children.
  - A male child has a 50 percent chance of getting the faulty gene and having the disorder because there is no back-up copy.
  - A female child, even if she gets a copy of the faulty gene from her mother, will receive another allele from her father.
- Red-green color blindness and hemophilia are examples of sex-linked inheritances.

## C. Chromosomal Abnormalities

- Chromosomal abnormalities typically occur because of errors in cell division.

- Klinefelter syndrome, found only in males, is caused by an extra female sex chromosome (shown by the pattern XXY).
- Turner syndrome results from a missing sex chromosome (XO) and is found only in females.
- Triple X syndrome results from an extra X chromosome. Also known as trisomy X, it is associated with delayed language and motor development and affects approximately 1 in 1,000 females.
- The most common genetic disorder in children is **Down syndrome**.
  - It is responsible for about 40 percent of cases of moderate-to-severe mental retardation as defined by performance on an intelligence test.
  - The condition is also called *trisomy-21* because it is characterized in more than 90 percent of cases by an extra 21st chromosome.
  - The most obvious physical characteristics associated with Down syndrome are distinct facial characteristics including a downward-sloping skin fold at the inner corners of the eyes.
  - Children with Down syndrome also tend to have slowed growth; poor muscle tone; congenital heart defects; thick hands; ear infections and early hearing loss; and impaired communication, language, memory, and motor skills.
- Approximately 1 in every 700 babies born alive has Down syndrome.
  - Although the risk of having a child with Down syndrome rises with age of the mother, because of the higher birthrates of younger women, there are actually more young mothers with children with Down syndrome.
- Rather than having the three familiar branching lines on their palm, children with Down syndrome are more likely to have one horizontal line across their palms, a characteristic known as the single transverse palmar crease.
  - This trait sometimes occurs in the general population, but it is more likely in children with Down syndrome.
- Children with Down syndrome, like other children with disabilities, tend to benefit cognitively, socially, and emotionally when placed in regular classrooms rather than in special schools and when given regular, intensive therapies to help them achieve important skills.

#### D. Genetic Counseling and Testing

- **Genetic counseling** can help prospective parents assess their risk of bearing children with genetic or chromosomal defects.
- People who have already had a child with a genetic defect, who have a family history of hereditary illness, who suffer from conditions known or suspected to be inherited, or

who come from ethnic groups at higher-than-average risk of passing on genes for certain diseases can get information about their likelihood of producing affected children.

- Screening for disorders can either happen before pregnancy, when parents can be screened for the presence of recessive genetic disorders, or after conception via genetic assessments such as chorionic villi sampling (CVS) and amniocentesis.
  - Both of these tests involve extracting fetal cells from the uterus, growing them in a laboratory, and doing genetic tests on them.
  - However CVS is generally done at 11 to 12 weeks gestation, while amniocentesis is done during the 16th week of pregnancy.
- Geneticists have made great contributions to the prevention of birth defects.

## IV. Studying the Influence of Heredity and Environment

### A. Measuring Heritability

- **Heritability** is a statistical estimate of how much heredity contributes to variations in a specific trait at a certain time within a given population.
- It does not refer to the relative influence of heredity and environment in a particular individual.
- It merely indicates the statistical extent to which genes contribute to a trait among a group of people.
- Heritability is expressed as a percentage ranging from 0.0 to 1.0; the higher the number, the greater the heritability of a trait.
- A heritability estimate of 1.0 indicates that genes are 100 percent responsible for variances in the trait within the population.
- Because heritability cannot be measured directly, researchers in behavioral genetics rely chiefly on three types of correlational research:
  - Family studies
  - Adoption studies
  - Twin studies
- These studies are all based on the assumption that if we know, on average, how many genes people share by virtue of knowing their genetic relationship, then we can measure how similar they are on traits (that is, their *concordance rate*) and work backward to determine the relative environmental influences.
  - For example, immediate family members are more genetically similar than more distant relatives, adopted children are genetically more like their biological families than their adoptive families, and monozygotic twins are more genetically similar than *dizygotic twins* (fraternal twins formed from two fertilized ova).

- In *family studies*, researchers measure the degree to which biological relatives share certain traits and determine whether or not the closeness of the familial relationship is associated with the degree of similarity.
  - In other words, the more closely two people are related, the more likely they will be similar on a trait if that trait is indeed genetically influenced.
  - Therefore, researchers use concordance rates on traits to infer genetic influences.
  - Generally, concordance rate is defined as the probability that two family members will share a trait.
- *Adoption studies* look at similarities between adopted children and their adoptive families and also between adopted children and their biological families.
  - When adopted children are more like their biological parents and siblings in a particular trait, we see the influence of heredity.
  - When they resemble their adoptive families more, we see the influence of environment.
- *Twin studies* compare pairs of monozygotic twins with same-sex dizygotic twins.
  - Monozygotic twins should be twice as genetically similar, on average, as dizygotic twins.
  - When monozygotic twins are more alike, or more *concordant*, on a trait than dizygotic twins, we see the likely effects of heredity.

## B. How Heredity and Environment Work Together

- The effects of genetic influences, especially on behavioral traits, are rarely inevitable.
- Even in a trait strongly influenced by heredity, the environment can have substantial impact.
- From conception, a combination of constitutional (biological and psychological), social, economic, and cultural factors help shape development.
  - The more advantageous these circumstances and the experiences to which they give rise, the greater the likelihood of optimum development.

### 1. Reaction Range and Canalization

- **Reaction range** is the conventional term for a range of potential expressions of a heredity trait.
  - Body size, for example, depends largely on biological processes, which are genetically regulated. Even so, a range of sizes is possible, depending on environmental opportunities and constraints, such as adequate nutrition and a person's own behavior.

- Heredity can influence whether a reaction range is wide or narrow.
  - In other words, the genotype places limits on the range of possible phenotypes.
- The metaphor of **canalization** illustrates how heredity restricts the range of development for some traits.
  - Some human characteristics, such as eye color, are so strongly programmed by genes they are said to be highly *canalized*; there is little opportunity for variance in their expression.
- Highly canalized traits require an extreme change in environment to alter their course.
  - For example, normal babies follow a predictable sequence of motor development: rolling, sitting, standing, walking, and running, in that order, at certain approximate ages.

## 2. Genotype–Environment Interaction

- **Genotype–environment interaction** usually refers to the effects of similar environmental conditions on genetically different individuals.
  - For example, many children are exposed to pollen and dust, but those with a genetic predisposition are more likely to develop allergic reactions.
  - Interactions can work the other way as well: Genetically similar children often develop differently depending on their home environments.
  - Thus it is the interaction of hereditary and environmental factors, not just one or the other, which produces certain outcomes.

## 3. Genotype–Environment Correlation

- The environment often reflects or reinforces genetic disorders. This tendency is called **genotype–environment correlation**, and it works in three ways to strengthen the phenotypic expression of a genotypic tendency:
  - *Active correlations*: Children actively select experiences that are consistent with their genetic tendencies. For example, a shy child is more likely than an outgoing child to spend time in solitary pursuits. This tendency to seek out environments compatible with one’s genotype is called *niche-picking*.
  - *Passive correlations*: Children not only inherit genes from their parents, they also inherit environments. For example, a musical parent is likely to create a home environment in which music is heard regularly, to give a child music lessons, and to take the child to musical events. If the child inherited the parent’s musical talent, the child’s musicality will reflect a combination of genetic and environmental influences. This type of correlation is called passive because the child does not

control it.

- *Reactive, or evocative, correlations*: Children with differing genetic makeup evoke different responses from adults. If a child shows interest and ability in music, parents who are not musically inclined may react by making a special effort to provide that child with musical experiences. This response, in turn, strengthens the child's genetic inclination toward music.

#### 4. What Makes Siblings So Different?

- You might assume that siblings, as they share approximately 50 percent of their genes, might be very similar to each other.
  - However, siblings can differ greatly in intellect and especially in personality, and this difference increases with age.
- One reason may be genetic differences, which lead children to need different kinds of stimulation or to respond differently to a similar home environment, and thus develop along increasingly divergent paths.
  - For example, twin studies have identified that genetic differences between siblings in part drive how family conflict is experienced.
- In addition, there are also **nonshared environmental effects** that result from the unique environment in which each child in a family grows up.
  - For example, parents and siblings may treat each child differently; a firstborn gets undivided attention, but laterborns must compete for it.

### V. Characteristics Influenced by Heredity and Environment

#### A. Physical and Physiological Traits

- Not only do monozygotic twins generally look alike, but they also are more concordant than dizygotic twins in their risk for medical disorders such as high blood pressure, heart disease, stroke, rheumatoid arthritis, peptic ulcers, and epilepsy.
- Another characteristic with a genetic basis is weight.
  - **Obesity** is measured by *body mass index*, or BMI (comparison of weight to height).
  - The risk of obesity is two to three times higher for a child with a family history of obesity.
  - However, this increased risk is not solely genetic. The kind and amount of food eaten in a particular home or in a particular social or ethnic group and the amount of exercise that is encouraged can increase or decrease the likelihood that a child will become overweight.

## B. Intelligence

- Heredity exerts a strong influence on general intelligence, as measured by intelligence tests, and a moderate effect on specific abilities such as memory, verbal ability, and spatial ability.
- While specific genes might contribute to intelligence, intelligence is influenced by the effects of large numbers of genes working together.
- Indirect evidence of the role of heredity in intelligence comes from adoption and twin studies.
  - Adopted children's scores on standardized intelligence tests are consistently closer to the scores of their biological mothers than to those of their adoptive parents and siblings; monozygotic twins are more alike in intelligence than dizygotic twins.
- Intelligence also depends in part on brain size and structure, which are under strong genetic control.
  - Experience counts, too; an enriched or impoverished environment can substantially affect the development and expression of innate ability.

## C. Temperament and Personality

- **Temperament** is a characteristic way of responding to the environment that is apparent from early infancy and is a precursor to personality.
  - Siblings tend to be similar in temperament.
- Scientists have identified genes directly linked with specific aspects of personality, such as a trait called *neuroticism*, which may contribute to depression and anxiety.

## D. Psychopathology

- There is evidence for a strong hereditary influence on such mental disorders as schizophrenia, autism, alcoholism, and depression.
  - All tend to run in families and to show greater concordance between monozygotic twins than between dizygotic twins.
- Schizophrenia is an example of heredity-environment interaction.
  - It is a neurological disorder that affects about 1 percent of the U.S. population each year.
  - It is characterized by loss of contact with reality; hallucinations and delusions; loss of coherent, logical thought; and inappropriate emotionality.
  - Estimates of heritability for this disorder are as high as 80 to 85 percent.



- However, monozygotic twins are not always concordant for schizophrenia, perhaps due to epigenesis.

## TEACHING AND LEARNING ACTIVITIES

### LECTURE TOPICS

#### Lecture Topic 2.1: Abnormal Sex Chromosome Patterns

Present the following conditions to illustrate chromosomal patterns.

**Klinefelter's syndrome:** Some males have an XXY pattern or an additional X chromosome. Physical characteristics include above-average height and long arms and legs. Other physical characteristics include possible breast development during puberty, a high-pitched voice, and light beard growth. Sometimes intellectual functioning is impaired and Klinefelter males are sterile.

**Turner's syndrome:** Some females are missing the Y chromosome and have an XO pattern. Physical characteristics include a short and immature appearance, webbed necks, eyelid fold, receding chins, and a broad chest. Supplemental estrogen therapy during adolescence does stimulate breast development and other secondary sex characteristics though Turner females are sterile. There is little or no impairment in intellectual ability and some show above-average IQ.

#### Lecture Topic 2.2: Down Syndrome

Since 1866, when Langdon Down first made medical records of what has sometimes been known as mongolism, much genetic, medical, and psychological research has focused on this syndrome. Although the exact cause of the chromosomal aberration responsible for the condition is not known, it is known that 97 percent of the cases are due to chance abnormalities during meiosis, the production of gametes. It is thought that, for some reason, chromosome 21 duplicates itself twice instead of once, does not divide properly when forming the gamete, or does not assemble into the correct cluster of chromosomes. As a result, an individual with Down syndrome receives a duplicate chromosome 21, resulting in a condition known as trisomy 21. Apparently this cause of Down syndrome is usually due to abnormalities in the mother's ovum. The syndrome occurs in all races equally (and in chimpanzees). As early as 10 years after Down's 1866 report, it was noted that mothers who are older when they bear children are more likely to have children with Down syndrome. Thus suspicion centers on aging effects and the meiotic process in females.

Before birth, the number of possible ova is fixed in the ovaries. In addition, meiosis has

partially occurred and is interrupted until just before ovulation. Thus a 45-year-old woman has ova that have been held in suspension at a very critical stage of chromosomal division for 45 years. Although the Down syndrome attributable to these causes cannot be traced through family histories or predicted in any other way, mothers who are older also have a greater recurrence risk (about three times the risk of a mother of the same age who has not had a child with Down syndrome). While the risk of having a child with Down syndrome rises with age of the mother, because of the higher birthrates of younger women, there are actually more young mothers with children with Down syndrome.

There is a second way in which Down syndrome occurs. It can be caused by genetic transmission by the sperm as well as the ovum. This accounts for the other 3 percent of affected children. Due to a structural abnormality in chromosomes called translocation, males as well as females can receive abnormal chromosome 21. They show no physical abnormalities. A karyotype of these individuals can reveal that they carry only 45 functioning chromosomes. When the sperm and ova are produced, some will contain duplicate chromosome 21 and thus increase the risk of having Down syndrome children.

Though not all affected children have the same physical characteristics, there are usually enough telltale characteristics to make an accurate syndrome diagnosis. The following are often present: epicanthic fold (a fold of skin above the tear duct); hypotonia (a relaxed condition of the muscles; often evident in the face); a large, furrow-surfaced tongue that often protrudes because the mouth cavity is smaller than normal; small stature, somewhat chubby; flat face with broad nose; stumpy hands that may have a straight crease across the palm (simian crease); irregular, abnormal set of teeth; sparse pubic hair; malformed ears; a gap between the first and second toes; and heart or other internal defects. Historically, many affected children would succumb to illness (usually respiratory infection) in the first year. Mainly due to antibiotics, most now survive until their late 20s and many until age 40 or 50 or older. Of the aging Down syndrome population, a large percentage has Alzheimer's disease, which has led to the discovery of genes on the 21st pair being linked to this disorder.

Today the Down syndrome child's most severe handicap is his/her mental retardation. Most individuals (60 to 70 percent) are either severely retarded (IQ = 25–49) or profoundly retarded (25–40 percent: IQ = 0–24). However, children with mosaic Down syndrome have normal or above-average intelligence and can perform school work adequately. It is reported that one exceptional individual became an admiral in the British navy. These children are typical in social/emotional intelligence. They show the same range of personality characteristics as neurotypical individuals. However, personality differences make each person unique. They benefit greatly from home care but can cause considerable strain on the family.

## **DISCUSSION TOPICS**

### **Discussion Topic 2.1: Infertility On The Rise?**

The first section of the chapter examines issues related to infertility and the technologies that have developed to manage it. Investigate with your class reasons for the rise in infertility in our present society. Has the declining birthrate and the rise in the number of couples who actively choose not to have children simply made us more sensitive to those couples with fertility problems? Were the estimates of infertility in the past inaccurately low because there were no services or advanced technology to help individuals and, therefore, no one spoke of their difficulties? Are there social and psychological factors that lead to infertility as well as the physical ones mentioned in the text? Is our society more stressful, and is fertility a result of this stress? Is there any relationship between changing gender roles for men and women and the rate of infertility?

### **Discussion Topic 2.2: The Availability Of Artificial Reproductive Technologies**

With infertility rates at their highest recorded levels, the demand for access to modern reproductive technologies is also great. Explore with your students the ethical and socioeconomic concerns surrounding this issue. Currently, the procedures are expensive. Should they be made available only to those with the ability to pay for them? How much coverage should be provided by medical insurance? Should access be limited only to childless offspring? Should single individuals be allowed to use the modern technologies to sire or bear offspring? Who is liable if the baby conceived by way of modern technology has a birth defect?

### **Discussion Topic 2.3: Prenatal Tests and Down Syndrome**

Until 2007, only pregnant women 35 and older were routinely tested to see if their fetuses had the extra chromosome that causes Down syndrome. However, under a new recommendation from the American College of Obstetricians and Gynecologists, doctors have begun to offer a new, safer screening procedure to all pregnant women, regardless of age.

Discuss with your students the pros and cons of the new screening procedure. Do you believe the parents should be given the chance to continue or terminate the pregnancy at their own free will? Should more efforts be undertaken to educate people about the value of accepting children with Down syndrome?

### **Discussion Topic 2.4: Obesity**

Science shows that genetics plays a role in obesity. Genes can directly cause obesity in disorders

such as Bardet-Biedl syndrome and Prader-Willi syndrome.

Ask your students to visit the following link and read through the article:

Discuss with your students the steps would-be parents can take to protect their children from obesity.

## **INDEPENDENT STUDIES**

### **Independent Study 2.1**

Select one of the genetic and chromosomal abnormalities mentioned in this chapter. Then, use the Internet to identify support groups or organizations for parents whose children have this particular abnormality.

## **KNOWLEDGE CONSTRUCTION ACTIVITIES**

### **Knowledge Construction Activity 2.1: Application of Terms**

This activity will use the principles of generative learning to assist students in gaining a better understanding of terms. Divide the class into groups of four or five. Assign each group the task of generating an example for a generative term from this chapter. The example that each group creates cannot be one that has been used in class or in the book. They must think of a new application for the term that they are given. Groups are allowed to use their books and notes. By creating their own example of the term, they demonstrate an understanding of the term to the level of application. There are several approaches that can be used in this exercise. Students may be given the entire list at once. Another strategy is to give all of the groups the same term to create an example and then go around the room to discuss outcomes. A third approach is to give each group a different term and see what examples they can generate.

#### **Some generative terms for Chapter 2**

Homozygous	Phenotype
Heterozygous	Genotype
Dominant inheritance	Natural selection
Recessive inheritance	Sex-linked inheritance
Epigenesis	Reaction range

### **Knowledge Construction Activity 2.2: Choosing Parenthood**

The decision to have a child is one of the most important decisions a person or couple can make. Consider some of the following questions in terms of the impact childbearing can have.

- Do I like doing the things parents generally do?
- Would I expect my child to take care of me in my old age?
- Could I find happiness in teaching and guiding a child, a teenager, or a young adult?
- Am I financially able to support a child?
- Would a child interfere with my freedom or educational plans?
- Would I be willing to devote a large part of my life to being a parent?
- Could I accept and love a child who was physically or mentally abnormal?

### **Knowledge Construction Activity 2.3: Infertility Options—Adoption**

Some couples choose not to have children because of the risk of genetic abnormalities. Other couples are infertile and cannot have children. Still others feel called to help children in need. Have students investigate the option of adoption. In your state, what are the legal requirements for adoption? Who can adopt a child? Are there standards for age, weight, health, sexual orientation, marital status, income and so on, of the parents? Can single persons adopt? Must there be a racial match between parent and child? What is the availability of infants, older children, minority-group children, and handicapped children?

### **Knowledge Construction Activity 2.4: Infertility Options And Embryo Rights**

A recent divorce case involved legal questions about frozen embryos. While married, the couple had attempted to have children. They finally availed themselves of modern reproductive techniques in which conception was achieved outside the womb. Several of the resulting embryos were frozen, awaiting the future needs of the couple. The legal questions focused on who had rights to the embryos when the marriage was dissolved. Assign students to research the topic and elicit responses as to which side they are on, and then make them argue for the opposite side.

### **Knowledge Construction Activity 2.5: Thinking About Genetic Testing**

If you were going to have a baby, would you want to know the sex of the child in advance? Genetic testing allows people to discover a great deal of information about their children before they are even born, but not everyone wants to know everything!

Assign three to four students to a group. Construct a set of situation cards for each group (3 x 5 index cards), place them facedown, and have each member choose one card.

**Situation cards:**

Your baby is likely to grow up to be very shy and anxious.

Your baby is likely to be very extroverted and hyperactive.

Your baby is likely to be autistic.

Your baby is likely to be a genius in math.

Your baby is likely to be a genius and neurotic (anxious).

Your baby is likely to be musically very talented but also likely to be poor at verbal tasks.

Your baby is likely to be just average.

Your baby is likely to be dyslexic.

Your baby is likely to grow up to be just like you.

Instruct participants to read the diagnosis aloud to their group and then comment on whether or not their child-rearing practices would change knowing this information in advance. Would they try to alter some outcomes more than others?

Further questions for discussion:

- What do you think people believe about genetic predisposition and the effect of the environment?
- What do you think about the value of prenatal diagnosis for the kinds of situations described in the cards?

**Knowledge Construction Activity 2.6: Gene-Environment Interaction**

The following exercise is to demonstrate the connection between passive, evocative, and active gene-environment correlation. First, assign students to three groups of four or five and assign each person to write down an example of a positive environmental factor that played a major role in the direction that his/her life took. Group One might look at the passive gene-environment correlation: association between the genotype a child inherits from her parents and the environment in which the child is raised. Group Two looks at evocative gene-environment correlations. This happens when an individual's (heritable) behavior evokes an environmental response. Group Three can look at the active gene-environment correlation. This occurs when an individual possesses a heritable propensity to select environmental exposure.

**Applied Activity 2.1: Gene-Environment Correlations (Class Discussion)**

“As you reflect upon your life to this point, it is possible to look back and see what factors have been influential in bringing you to where you are today. For each of the following terms, give an example of an environmental influence that may have been linked by your genetics. An issue to

consider might be what has led you to choose the career path that you have selected (Nursing, Education, etc.).”

1. Passive Genotype-Environmental Correlation.
2. Reactive or Evocative Genotype-Environmental Correlation.
3. Active Genotype-Environmental Correlation.

### Applied Activity 2.2: Genetics (Class Discussion)

Do an Internet search on the term “genetics.” Visit several websites to answer the following questions:

- What constitutes the study of genetics?
- Who might benefit from genetic counseling?
- What kind of information do you find on the Internet?
- Are there agencies, universities, or medical centers who are studying human genetics?
- Are there certification requirements for genetic counselors?
- What are some of the findings of genetics research?

### The Ten-Minute Test

Name: \_\_\_\_\_

Answer the questions below utilizing the following terms:

clone	heritability	zygote
reaction range	chromosomes	genes
heterozygous	dominant	recessive
phenotype	monozygotic	genotype
sex-linked inheritance	behavioral genetics	fertilization
dizygotic	homozygous	epigenesis

1. A one-celled organism resulting from fertilization is called a\_\_\_\_\_.
2. Twins conceived by the union of two different ova with two different sperm are called \_\_\_\_\_twins.
3. Small segments of DNA located in definite positions on particular chromosomes are called\_\_\_\_\_.

4. \_\_\_\_\_ means that an organism possesses two identical alleles for a trait.
5. \_\_\_\_\_ inheritance is when a child receives contradictory alleles and only one is expressed.
6. \_\_\_\_\_ is the observable characteristics of a person.
7. A pattern of inheritance in which certain characteristics carried on the X chromosome inherited from the mother are transmitted differently to her male and female offspring is called \_\_\_\_\_.
8. \_\_\_\_\_ is the quantitative study of relative heredity and environmental influences.
9. \_\_\_\_\_ is a statistical estimate of the contribution of heredity to individual differences in a specific trait within a given population.
10. \_\_\_\_\_ is the potential variability in the expression of a hereditary trait.

### **Answers to the Ten-Minute Test**

1. zygote
2. dizygotic
3. genes
4. homozygous
5. dominant
6. phenotype
7. sex-linked inheritance
8. behavioral genetics
9. heritability
10. reaction range

### **Resources for Instructors**

#### **Books and Journal Articles**

*Am I Parent Material?* National Organization for Non-Parents, 3 North Liberty Street, Baltimore, MD 21201



Huxley, A. (1946). *Brave new world*. New York: Harper and Row.

Moldin, S.O. & Rubenstein, R.R.L. (2006). *Understanding autism: From basic neuroscience to treatment*. Boca Raton, Florida: Taylor & Francis.

Nussbaum, R.L., McInnes, R.R., & Willard, H.F. (2007). *Thompson & Thompson genetics in medicine*. Philadelphia: Pennsylvania: Saunders.

Pennington, Moon, Edgin, Stredron, & Nadel. (2003). The neuropsychology of Down syndrome. *Child Development*, 74, 75-93.

Plomin, R. (2004). *Nature and nurture: An introduction to human behavioral genetics*. Wadsworth.

## **Internet Resources**

Genetic Science Learning Center. This site provides downloadable teaching tips and classroom resources covering topics such as basic genetics, genetic disorders, and genetic testing.

Human Genome project.. This site provides the latest information on uncovering all of the genes in the human cell.

National Down Syndrome Society. site provides information and support.

University of Kansas Medical Center, Genetics Education Center. Web address: site is an excellent companion resource to the Human Genome project with exhibits, links, and articles.