Beginnings: A Preview

PART 2
Forming A New Life: Conception, Heredity, and Environment

Chapter 3
Guideposts for Study

1. How Does Conception Normally Occur, and How Have Beliefs About Conception Changed?

2. What Causes Infertility, and What Are Alternative Ways of Becoming Parents?
Guideposts for Study

- 3. What Genetic Mechanisms Determine Sex, Physical Appearance, and Other Characteristics?

- 4. How Are Birth Defects and Disorders Transmitted?
5. How Do Scientists Study the Relative Influences of Heredity and Environment, and How Do Heredity and Environment Work Together?

6. What Roles Do Heredity and Environment Play in Physical Health, Intelligence, and Personality?
Becoming Parents: How Conception Occurs

- **Changing Theories of Conception**
  - Animaliculists believed that humans were pre-formed in head of sperm
  - Ovists believed that humans were pre-formed in eggs and activated by sperm
  - Kaspar Friedrich Wolff demonstrated that embryos are not pre-formed and are contributed to by both parents equally supporting a theory of the gradual building of structures
Becoming Parents:
How Conception Occurs

How Fertilization Takes Place

- Sperm and ovum— the male and female gametes (sex cells)— combine to create a zygote
- Zygote will duplicate itself by cell division to create a baby
Becoming Parents: How Conception Occurs

- **How Fertilization Takes Place**
  - Girls have about 2 million ova in their ovaries at birth, each contained in a follicle.
  - During ovulation, a mature follicle is ruptured and expelled from the ovary.
  - Ovum is swept through the fallopian tube toward the uterus.
Becoming Parents: How Conception Occurs

How Fertilization Takes Place

- Several hundred million sperm are produced in the testicles each day.
- Sperm enter the vagina through ejaculation and attempt to reach the cervix; eventually a few will arrive in the fallopian tubes where fertilization takes place.
Infertility

- Defined as an inability to conceive a baby after 12 months of trying
- 7 percent of U.S. couples are infertile
- Women’s fertility begins to decline in the late 20s
- Men’s fertility begins to decline in the late 30s
Infertility

- Causes of Infertility
  - Men
    - Too few sperm; 60-200 per ejaculation is too low
    - Duct may be blocked, or sperm are unable to “swim” well enough
Infertility

- **Causes of Infertility**
  - **Women**
    - Blockage in the fallopian tube
    - Failure to produce ova or good quality ova
    - Mucus in the cervix
    - Disease in the uterine lining
<table>
<thead>
<tr>
<th>Condition</th>
<th>Explanations</th>
<th>Treatments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Impaired delivery of sperm into vagina</td>
<td>Sexual problems, including erectile dysfunction, premature ejaculation, and painful intercourse. Physical problems, including failure to produce semen, blockage of ejaculatory ducts, other structural defects, and antibodies that weaken or disable sperm.</td>
<td>Sexual problems can be treated with medication or behavioral therapy. Physical problems may require surgery. Assisted reproduction techniques may include in vitro fertilization, electrical stimulation of ejaculation, or surgical retrieval of sperm (if blockage is present).</td>
</tr>
<tr>
<td>Age</td>
<td>Gradual decline in fertility, commonly in men older than 35.</td>
<td>Correct health and lifestyle problems if possible.</td>
</tr>
<tr>
<td>General health and lifestyle issues</td>
<td>Emotional stress, malnutrition, obesity, alcohol and drugs, tobacco smoking, cancer treatments, severe injury, surgery, and other medical conditions may impair sperm production.</td>
<td>Avoid unhealthy exposures.</td>
</tr>
<tr>
<td>Environmental exposure</td>
<td>Overexposure to heat (in saunas or hot tubs), toxins, and certain chemicals, such as pesticides, lead, and chemical solvents.</td>
<td>Avoid unhealthy exposures.</td>
</tr>
<tr>
<td>Female Causes</td>
<td>Most frequent cause: inflammation of the fallopian tube due to chlamydia, a sexually transmitted disease; tubal damage with scarring may result in an ectopic pregnancy, in which the fertilized egg is unable to pass through the fallopian tube and implant in the uterus. Other causes: benign uterine fibroid tumors and pelvic adhesions (bands of scar tissue) formed after pelvic infections, appendicitis, or pelvic or abdominal surgery.</td>
<td>Laparoscopic surgery to repair or open fallopian tubes; in vitro fertilization.</td>
</tr>
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<td>Fallopian tube damage or blockage</td>
<td>Uterine tissue implanted outside the uterus can lead to scarring and inflammation, which may prevent transfer of ovum to fallopian tube and cause pelvic pain. Ovarian cysts.</td>
<td>Ovulation therapy (medication to stimulate ovulation) or in vitro fertilization.</td>
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<tr>
<td>Endometriosis</td>
<td>Uterine tissue implanted outside the uterus can lead to scarring and inflammation, which may prevent transfer of ovum to fallopian tube and cause pelvic pain. Ovarian cysts.</td>
<td>Ovulation therapy (medication to stimulate ovulation) or in vitro fertilization.</td>
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<tr>
<td>Ovulation disorders</td>
<td>Any condition that prevents the release of a mature ovum from the ovary. Specific causes include hormonal deficiencies, injury to the hypothalamus or pituitary gland, pituitary tumors, excessive exercise, and eating disorders.</td>
<td>Fertility drugs.</td>
</tr>
<tr>
<td>Polycystic ovary syndrome</td>
<td>Increase in production of the hormone androgen can prevent production of mature ovum. Common symptoms are absent or infrequent menstruation; dark or thick hair on chin, upper lip, or abdomen; acne; and oily skin.</td>
<td>Fertility drugs, particularly clomiphene.</td>
</tr>
<tr>
<td>Early menopause</td>
<td>Ovarian failure before age 35 may be associated with autoimmune disease, hypothyroidism (too little thyroid hormone), radiation or chemotherapy for cancer treatment, or tobacco smoking.</td>
<td>In vitro fertilization with donated ova.</td>
</tr>
</tbody>
</table>


Table 3-1
Infertility

- Treatments For Infertility
  - Hormone treatment
  - Drug therapy
  - Surgery
Infertility

Alternative Ways to Parenthood

Assisted Reproduction Technology (ART)

- In Vitro Fertilization (IVF)
  - Increases the likelihood of multiple, usually premature, births

- In Vitro Maturation (IVM)
  - Performed earlier in the monthly cycle
  - Makes hormone injections unnecessary
  - Diminishes the likelihood of multiple births
DNA is the genetic material in all living cells. It consists of four chemical units, called bases. These bases are the letters of the DNA alphabet. A (adenine) pairs with T (thymine) and C (cytosine) pairs with G (guanine). There are 3 billion base pairs in human DNA.

Letters of the DNA alphabet

T = Thymine
A = Adenine
G = Guanine
C = Cytosine
Infertility

- Alternative Ways to Parenthood
  - Male infertility
    - Intracytoplastic sperm injection (ICSI)
    - Artificial insemination
    - Artificial insemination by a donor (AID)
Infertility

- Alternative Ways to Parenthood
  - Gamete intrafallopian transfer (GIFT)
  - Zygote intrafallopian transfer (ZIFT)
  - Surrogate motherhood
Multiple Births

- Dizygotic twins – fraternal twins
- Monozygotic twins – identical twins
- Semi-identical twins
- The incidence of multiple births has elevated rapidly since 1980
  - Suleman & Chukwu examples
  - Risks during pregnancy and delivery of multiple birth
Mechanisms of Heredity

■ The Genetic Code
  ■ Deoxyribonucleic acid (DNA)
    ■ Bases – Letters of the genetic code which cellular machinery “reads”
      ■ Adenine (A)
      ■ Thymine (T)
      ■ Cytosine (C)
      ■ Guanine (G)
Mechanisms of Heredity

- **The Genetic Code**
  - **Chromosomes**
  - **Genes**
    - Each is located in a definite position on its chromosome and contains thousands of base pairs
    - Every cell (except sex cells) has 23 pairs of chromosomes—46 in all
    - Monomorphic
    - Polymorphic
Father has an X chromosome and a Y chromosome. Mother has two X chromosomes. Male baby receives an X chromosome from the mother and a Y chromosome from the father. Female baby receives X chromosomes from both mother and father.
Mechanisms of Heredity

What Determines Sex?
- Father’s sperm determines gender
- Ovum provides X chromosomes
- Sperm contains either X or Y chromosomes
- Gene for maleness, SRY gene
Mechanisms of Heredity

- Patterns of Genetic Transmission
  - Every offspring gets a pair of alleles for each characteristic, one from each parent
  - When an offspring receives alleles for two contradictory traits, only one of them, the dominant one, shows itself—a heterozygous characteristic
Mechanisms of Heredity

- **Patterns of Genetic Transmission**
  - The expression of a recessive trait occurs only when a person receives the recessive allele from both parents.
  - A *homozygous* characteristic results when both alleles are the same.
Dominant and Recessive Inheritance

Mother: Dd
Father: Dd

Offspring:
- DD
- Dd
- Dd
- dd
Mechanisms of Heredity

- **Polygenic inheritance** involves the interaction of several genes
- **Molecular genetics** identifies specific genes linked to particular behavioral traits
- **Multifactorial transmission**: combination of genetic and environmental factors
  - Genotype and phenotype
Epigenesis

- Defined as a mechanism that turns genes on or off to affect how those genes are expressed
  - Suggests a bidirectional interplay with nongenetic influences
  - Determines the functions of body cells
  - Cells are particularly susceptible to epigenetic modification during critical periods such as puberty and pregnancy
Genetic and Chromosomal Abnormalities

- Dominant or Recessive Inheritance of Defects
  - Some abnormal traits are dominant
    - Achondroplasia (type of dwarfism)
    - Huntington’s disease
  - Some abnormal traits are recessive in ethnic groups
    - Tay-Sachs
    - Sickle-cell anemia
<table>
<thead>
<tr>
<th>Pattern/Name</th>
<th>Characteristics*</th>
<th>Incidence</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>XYY</td>
<td>Male; tall stature; tendency to low IQ, especially verbal.</td>
<td>1 in 1,000 male births</td>
<td>No special treatment</td>
</tr>
<tr>
<td>XXX (triple X)</td>
<td>Female; normal appearance, menstrual irregularities, learning disorders, mental retardation.</td>
<td>1 in 1,000 female births</td>
<td>Special education</td>
</tr>
<tr>
<td>XXY (Kleinfelter)</td>
<td>Male; sterility, underdeveloped secondary sex characteristics, small testes, learning disorders.</td>
<td>1 in 1,000 male births</td>
<td>Hormone therapy, special education</td>
</tr>
<tr>
<td>XO (Turner)</td>
<td>Female; short stature, webbed neck, impaired spatial abilities, no menstruation, infertility, underdeveloped sex organs, incomplete development of secondary sex characteristics.</td>
<td>1 in 1,500 to 2,500 female births</td>
<td>Hormone therapy, special education</td>
</tr>
<tr>
<td>Fragile X</td>
<td>Minor-to-severe mental retardation; symptoms, which are more severe in males, include delayed speech and motor development, speech impairments, and hyperactivity; the most common inherited form of mental retardation.</td>
<td>1 in 1,200 male births; 1 in 2,000 female births</td>
<td>Educational and behavioral therapies when needed</td>
</tr>
</tbody>
</table>

*Not every affected person has every characteristic.
Genetic and Chromosomal Abnormalities

- **Sex-Linked Inheritance of Defects**
  - Sex-linked inheritance
    - Certain recessive disorders are linked to genes on the sex chromosomes
    - Male and female children affected differently
    - X chromosomes - Mother is carrier
      - Can be overridden
    - Y chromosome – Father is carrier
      - Cannot be overridden
Genetic and Chromosomal Abnormalities

- **Sex-Linked Inheritance**
  - 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
Genetic and Chromosomal Abnormalities

- Chromosomal Abnormalities
  - Errors in cell division
  - Extra or missing chromosome
  - Klinefelter syndrome XXY
  - Turner syndrome XO
  - Down syndrome
<table>
<thead>
<tr>
<th>Problem</th>
<th>Characteristics of Condition</th>
<th>Who Is at Risk</th>
<th>What Can Be Done</th>
</tr>
</thead>
<tbody>
<tr>
<td>Alpha, antitrypsin deficiency</td>
<td>Enzyme deficiency that can lead to cirrhosis of the liver in early infancy and emphysema and degenerative lung disease in middle age.</td>
<td>1 in 1,000 white births</td>
<td>No treatment.</td>
</tr>
<tr>
<td>Alpha thalassemia</td>
<td>Severe anemia that reduces ability of the blood to carry oxygen; nearly all affected infants are stillborn or die soon after birth.</td>
<td>Primarily families of Malaysian, African, and Southeast Asian descent</td>
<td>Frequent blood transfusions.</td>
</tr>
<tr>
<td>Beta thalassemia (Cooley’s anemia)</td>
<td>Severe anemia resulting in weakness, fatigue, and frequent illness; usually fatal in adolescence or young adulthood.</td>
<td>Primarily families of Mediterranean descent</td>
<td>Frequent blood transfusions.</td>
</tr>
<tr>
<td>Cystic fibrosis</td>
<td>Overproduction of mucus, which collects in the lung and digestive tract; children do not grow normally and usually do not live beyond age 30; the most common inherited lethal defect among white people.</td>
<td>1 in 2,000 white births</td>
<td>Daily physical therapy to loosen mucus; antibiotics for lung infections; enzymes to improve digestion; gene therapy (in experimental stage).</td>
</tr>
<tr>
<td>Duchenne muscular dystrophy</td>
<td>Fatal disease usually found in males, marked by muscle weakness; minor mental retardation is common; respiratory failure and death usually occur in young adulthood.</td>
<td>1 in 3,000 to 5,000 male births</td>
<td>No treatment.</td>
</tr>
<tr>
<td>Hemophilia</td>
<td>Excessive bleeding, usually found in males; in its most severe form, can lead to crippling arthritis in adulthood.</td>
<td>1 in 10,000 families with a history of hemophilia</td>
<td>Frequent transfusions of blood with clotting factors.</td>
</tr>
<tr>
<td>Neural-tube defects Anencephaly</td>
<td>Absence of brain tissues; infants are stillborn or die soon after birth.</td>
<td>1 in 1,000</td>
<td>No treatment.</td>
</tr>
<tr>
<td>Spina bifida</td>
<td>Incompletely closed spinal canal, resulting in muscle weakness or paralysis and loss of bladder and bowel control; often accompanied by hydrocephalus, an accumulation of spinal fluid in the brain, which can lead to mental retardation.</td>
<td>1 in 1,000</td>
<td>Surgery to close spinal canal prevents further injury; shunt placed in brain drains excess fluid and prevents mental retardation.</td>
</tr>
<tr>
<td>Phenylketonuria (PKU)</td>
<td>Metabolic disorder resulting in mental retardation.</td>
<td>1 in 15,000</td>
<td>Special diet begun in first few weeks of life can prevent mental retardation.</td>
</tr>
<tr>
<td>Polycystic kidney disease</td>
<td>Infantile form: enlarged kidneys, leading to respiratory problems and congestive heart failure. Adult form: kidney pain, kidney stones, and hypertension resulting in chronic kidney failure.</td>
<td>1 in 1,000</td>
<td>Kidney transplants.</td>
</tr>
<tr>
<td>Sickle-cell anemia</td>
<td>Deformed, fragile red blood cells that can clog the blood vessels, depriving the body of oxygen; symptoms include severe pain, stunted growth, frequent infections, leg ulcers, gallstones, susceptibility to pneumonia, and stroke.</td>
<td>1 in 500 African Americans</td>
<td>Painkillers, transfusions for anemia and to prevent stroke, antibiotics for infections.</td>
</tr>
<tr>
<td>Tay-Sachs disease</td>
<td>Degenerative disease of the brain and nerve cells, resulting in death before age 5.</td>
<td>Historically found mainly in eastern European Jews</td>
<td>No treatment.</td>
</tr>
</tbody>
</table>

Source: Adapted from AAP Committee on Genetics, 1996; NIH Consensus Development Panel, 2001; Tisdale, 1985, pp. 68-69.
Genetic and Chromosomal Abnormalities

- **Genetic Counseling and Testing**
  - Prospective parents assess mathematical risk of bearing children with genetic or chromosomal defects
  - *Karyotype*
  - Genetic determinism
Nature and Nurture: Influences of Heredity and Environment

- **Studying the Relative Influences of Heredity and Environment**
  - **Behavioral genetics** seeks to measure how much heredity and environment influence a particular trait
  - **Heritability** – a statistical estimate of how heredity influences diversity within a specific trait in a given population
Nature and Nurture: Influences of Heredity and Environment

- Studying the Relative Influences of Heredity and Environment
  - If heredity influences a particular trait:
    - Siblings should be more alike than cousins
    - Monozygotic twins should be more alike than dizygotic twins (concordance)
    - Adopted children should be more like biological than adoptive parents
Nature and Nurture: Influences of Heredity and Environment

- How Heredity and Environment Work Together
  - Genes and experience form part of a complex developmental system
  - Reaction Range
  - Canalization
Figure 3-8

Heritability = 100%

Uniform lighting

Differences between groups is totally environmental

Uniform nutrient solution: normal

Uniform nutrient solution: deficient
Nature and Nurture: Influences of Heredity and Environment

- How Heredity and Environment Work Together
  - Genotype-Environment Interaction
  - Genotype-Environment Correlation
    - Passive
    - Reactive or evocative
    - Active
Nature and Nurture: Influences of Heredity and Environment

- How Heredity and Environment Work Together
  - Niche-picking tendency
  - Nonshared environmental effects
  - Effects of experience on development: interactions of parenting, nonfamilial influences, and broader context in which families live
Some Characteristics Influenced by Heredity and Environment

- Physical and Physiological Traits
  - Obesity
- Intelligence
- Personality
  - Temperament
- Psychopathology
  - Schizophrenia