Part I  Chapter Three

Heredity and Environment

- The Genetic Code
- Sex, Multiple Births, and Fertility
- From One Cell to Many
- Genotype and Phenotype
- Chromosomal and Genetic Abnormalities
- Genetic Counseling and Testing
The Genetic Code

Genes affect every aspect of your development and come from your parents and will be passed on to your children.

Genes play a leading role in the drama of human development, yet they rarely take center stage. Genes are pervasive and powerful, but they are also hidden and elusive.
• **deoxyribonucleic acid (DNA):** molecule that contains the chemical instructions for cells to manufacture various proteins

• **chromosome:** one of 46 molecules of DNA (in 23 pairs) that each cell of the human body contains and that, together, contain all the genes

• **gene:** section of chromosome and the basic unit for the transmission of heredity, consisting of a string of chemicals that are instructions for the cell to manufacture certain proteins
CELL

NUCLEUS

23 pairs of chromosomes

CHROMOSOME
(DNA MOLECULE = DOUBLE HELIX)

Gene

Gene

Gene

Gene

PROTEIN

Triplet (specifies an amino acid)

Triplet (specifies an amino acid)

Strands of double helix

Amino acid

Amino acid
What Genes Are

• **allele**: any of possible forms in which a gene for a particular train can occur

• **genome**: full set of genes that are the instructions to make an individual member of a certain species
The Beginnings of Life

- **gamete**: a reproductive cell; sperm or ovum that can produce a new individual if it combines with a gamete from the other sex to make a zygote.

- **zygote**: the single cell formed from the fusing of two gametes, a sperm and an ovum.

- **genotype**: organism’s entire genetic inheritance, or genetic potential.
The Moment of Conception  

Mapping the Karyotype
Sex, Multiple Births, and Fertility

• The basics of chromosomes and genes hold true for all living creatures.
  – **23rd pair**: chromosome pair that determines zygote’s sex
  – other 22 pairs are autosomes, the same whether the 23rd pair is for a male or a female
Male or Female?

**XX is a 23rd chromosome pair** that consists of two X-shaped chromosomes, one each from the mother and the father.

XX zygotes become females.

**XY is a 23rd chromosome pair** that consists of an X-shaped chromosome from the mother and a Y-shaped chromosome from the father.

XY zygotes become males.
Determining a Zygote’s Sex

Possible Combinations of Sex Chromosomes

Father’s chromosomes 44+XY

Mother’s chromosomes 44+XX

Sperm

22+ Y

22+ Y

22+ X

22+ X

Ova

22+ X

22+ X

22+ X

22+ X

Zygotes

44+ XY

44+ XY

44+ XX

44+ XX

Male

Male

Female

Female
Twins

- **monozygotic (MZ) twins**: twins from one zygote
- **other monozygotic multiple births can occur**
• **dizygotic (DZ) twins**: two separate ova fertilized by two separate sperm at roughly the same time (also called *fraternal twins*)
Assisted Reproduction

- **infertility**: inability to conceive after at least a year of trying to do so via sexual intercourse

- **assisted reproductive technology (ART)**: general term for techniques designed to help infertile couples conceive and then sustain a pregnancy
• **in vitro fertilization (IVF):** fertilization that takes place outside woman’s body involves mixing sperm with ova that have been surgically removed from the woman’s ovary
  – one-third of all **IVF cycles** produce a pregnancy

• some infertile adults may achieve conception and birth with the assistance of:
  – donated sperm
  – donated ova
  – donated womb
• Complications and birth defects may increase with IVF
  – when several zygotes are implanted at once low-birthweight is a common complication

• In the U.S. because of ART:
  – triplet births increased by 500 percent since 1970
  – twins birth have almost doubled in this time
From One Cell to Many

• **Genotype:**
  – When sperm and ovum combine into zygote they establish all genes that the developing person will have.

• **Phenotype**
  – Genes that are either ignored or amplified on the genotype. This is actually the observable characteristic of a person, including appearance, personality, intelligence, and all other traits.
New Cells, New Functions

• Within hours after conception, the zygote begins duplication and division.
• The 23 pairs of chromosomes duplicate, forming two complete sets of the genome.
• The two sets move toward opposite sides of the zygote; the single cell splits.
• Each cell contains the entire original genetic code.
• At birth zygote has become about 10 trillion cells.
  – by adulthood more than 100 trillion cells
    • no matter how large the total number
    • no matter how much division and duplication occur

• almost every cell carries exact copy of complete genetic instructions inherited by one-celled zygote
Stem Cells

• **Stem cells** are able to produce any type of cell.
  – Result of differentiation and specialization.
    • At the eight-cell stage (approx.) differentiation occurs. Cells now begin to specialized.
    • Specialization causes some to be part of an eye, others part of a finger.
Gene-Gene Interactions

• Many new discoveries have followed the completion of the Human Genome Project in 2001.

• The Project discovered:
  – Humans have far fewer than the 100,000 genes previously thought.
  – It is not always easy to figure out where one gene starts and another ends.
  – There is similarity of the genes of all living creatures.
Additive Heredity

- Some alleles are called **additive genes** because their effects add up to influence the phenotype.

- **additive gene**: gene that has several alleles, each of which contributes to the final phenotype
Dominant-Recessive Heredity

• **dominant-recessive pattern:** interaction of a pair of alleles in such a way that the phenotype reveals the influence of one allele (the dominant gene) more than that of the other (the recessive gene)

• special case of the dominant-recessive pattern occurs with genes that are x-linked, located on the x chromosome
• **carrier:** a person whose genotype includes a gene not expressed in phenotype
  
  – unexpressed gene occurs in half of the carrier’s gametes
  
  – generally only when inherited from both parents does characteristic appear in the phenotype
More Complications

• At the beginning of the twentieth century, enthusiasm for genetics led to eugenics.  
  – literally, “good genes”  
  – the idea that selective breeding was needed to improve the human race.  
  • led to programs of forced sterilization of people who were thought to have “bad genes”
Genotype and Phenotype

1. Genes affect every aspect of human behavior, including social and cognitive behavior.

2. Most environmental influences on children raised in the same home are not shared.

3. Each child’s genes elicit other people’s responses, and these responses shape development. In other words, a child’s environment is partly the result of his or her genes.

4. Children, adolescents, and especially adults choose environments that are compatible with their genes (called niche-picking), and thus genetic influences increase with age.
Alcoholism

• drug addiction and alcoholism
  – Was once considered to be….
    • considered moral weakness
    • social ill
    • personality defect
    • some combination of the three

• inherited biochemistry makes people vulnerable to various addictions
  – genes create addictive pull that can be:
    • overpowering
    • extremely weak
    • somewhere in between
• personality traits that encourage drinking and drug taking:
  – quick temper
  – sensation seeking
  – high anxiety

• gender mitigates or increases susceptibility
Visual Acuity

• Age and genes are powerful influences on vision.
  – Newborns cannot focus more than 2 feet.
  – Children see better each year until about age 8.
  – Many adolescents develop nearsightedness (myopia) when their eyeball shape changes.
Heritability of Nearsightedness

- **heritability**: statistic that indicates what percentage of the variation in a particular trait can be traced to genes:
  - within a particular population
  - in a particular context and era
International Variations

• In some places, visual problems are caused almost entirely by hazards in the environment.
  – In many African nations, heritability of visual acuity is close to zero.
  – Nutrition, not genes, is why some children see better.

• In Singapore, Taiwan, and Hong Kong:
  – The surge in childhood myopia has been called an epidemic.
  – First published research on this phenomenon appeared in 1992.
  – Scholars noticed significant change in the results of army-mandated medical exams of all 17-year-olds males in Singapore.
Chromosomal and Genetic Abnormalities

• abnormalities caused by identifiable problems, such as extra chromosome or a single gene
  – study of these problems relevant to the study of development because:
    • they provide insight into the complexities of nature and nurture
    • knowing their origins helps limit effects
    • information combats surrounding prejudice
Down Syndrome

• condition where person has 47 chromosomes instead of usual 46
  – Three, rather than two, chromosomes at 21st position

• Adults with down syndrome age faster than other adults, with ailments of aging usually setting in at about age 30.
Abnormalities of the 23\textsuperscript{rd} Pair

- humans have at least 44 autosomes and one X chromosome
  - zygote cannot develop without those 45

- About 1 of every 500 infants has only one X and no Y.
  - embryo cannot develop without an X chromosome
  - odd number of X chromosomes impairs:
    - cognition
    - psychosocial development
    - sexual maturation

- If a child has three sex chromosomes instead of two he/she may
  - seem normal until puberty
Gene Disorders

- Everyone carries genes or alleles that could produce serous diseases or handicaps in the next generation.
  - most genes contribute only small amount to a disorder
  - exact impact of each allele implicated in multifactorial disorder not yet known
Fragile X Syndrome

- genetic disorder in which part of the X chromosome seems to be attached by very thin string of molecules
- cause is single gene that has more than 200 repetitions of one triplet
  - Inherited mental retardation
  - Emotional problems
  - Poor social skills
Recessive-Gene Disorder

• Most recessive disorders are on autosomes that are not X-linked.
  – examples:
    • cystic fibrosis
    • thalassemia
    • sickle-cell anemia
  – sometimes a person who carried a lethal gene has many descendants who marry each other
  – genetic disease becomes common in that group
Genetic Counseling and Testing

- **Genetic counseling**: consultation and testing to learn about one’s genetic heritage, including harmful conditions that may pass along to any children they may conceive.
Who Should Get Counseling, and When?

• individuals and couples who:
  – have a parent, sibling, or child with a serious genetic condition
  – have had miscarriages or stillbirths
  – are infertile
  – are from the same ethnic group, particularly if related to each other
  – women over age 35 and men over age 40
• genetic counselors, scientists, and general public usually favor testing
  – is some information better than none?

• high risk individuals do not always want to know
  – truth might jeopardize marriage, insurance coverage, or chance of parenthood
Is Knowledge Always Power?

- phenylketonuria (PKU): genetic disorder in which child’s body unable to metabolize an amino acid phenylalanine
  - Resulting buildup of phenylalanine in body fluids causes brain damage, progressive mental retardation, and other symptoms.