Chapter 2

GENETICS
AND PRENATAL DEVELOPMENT
Figure 2.2   The Chemical Structure of DNA

DNA is composed of nucleotide pairs.
Human Genome

- 23,000 genes in our 46 chromosomes
- 3 billion nucleotide Pairs
  - adenine + thymine
  - guanine + cytosine

What is the sex of this person?

**Figure 2.1  The Human Genome**
The 46 chromosomes in the human genome are organized into 23 pairs.
Genetic Basics
Expression of Traits

• **Genotype**- The totality of an individual’s genes
• **Phenotype**- Actual Characteristics
• Environment can affect which genotype is expressed.

Ex: born w/a genotype that included exceptional music ability but you were never exposed to music and instruments.

LO 2.1 Genotype and Phenotype
• **Dominant Genes**-Expressed characteristics influences the phenotype  
• **Recessive Genes**-Not expressed, although it is a part of the genotype (genetic background)  
• **Allele**-Alternate form of a gene. Has a different nucleotide base sequence.  
• **Dominant-Recessive Inheritance**- a pair of chromosomes each has 1 dominant and 1 recessive gene.
# Table 2.1 Traits With Single-Gene Dominant–Recessive Inheritance

<table>
<thead>
<tr>
<th>Dominant</th>
<th>Recessive</th>
</tr>
</thead>
<tbody>
<tr>
<td>Curly hair</td>
<td>Straight hair</td>
</tr>
<tr>
<td>Dark hair</td>
<td>Blonde hair</td>
</tr>
<tr>
<td>Facial dimples</td>
<td>No dimples</td>
</tr>
<tr>
<td>Normal hearing</td>
<td>Deafness (some forms)</td>
</tr>
<tr>
<td>Normal vision</td>
<td>Nearsighted vision</td>
</tr>
<tr>
<td>Freckles</td>
<td>No freckles</td>
</tr>
<tr>
<td>Unattached ear lobe</td>
<td>Attached ear lobe</td>
</tr>
<tr>
<td>Can roll tongue in U-shape</td>
<td>Cannot roll tongue</td>
</tr>
</tbody>
</table>
• The 23rd chromosomes pair determines sex
• Males more vulnerable to X-linked recessive disorders

Figure 2.4  X-Linked Inheritance in Hemophilia
Why are males more vulnerable to recessive disorders carried on the X chromosome?
Genetic Basics
Genes and Environment

• Behavior Genetics- field that aims to identify the extent to which genes influence behavior.
• twins studies & adoption studies to see the influences of genes
• Monozygotic twins = 100% of genes in common
• Dizygotic twins = 40-60% of genes in common.

LO 2.3 Behavior Genetics
Genetic Basics
Genes and Environment

• **Heritability** - An estimate of the extent to which genes are responsible for differences among persons within a specific population with values ranging from 0-1.00
  - ex: intelligence estimates for children & teens = .50
  - ex: intelligence increases from childhood to adulthood but your genes don't change...

so what does?

LO 2.3 Behavior Genetics
Concordance Rates- Percentage that indicates degree of similarity in phenotype among pairs of family members. Rates range from 0%-100%

- Studies made by comparing MZ & DZ twins

Ex: concordance rates for schizophrenia

**MZ twins** = 50% *(when 1 twin has schizophrenia, 50% of the time the other twin has schizophrenia)*

**DZ twins** = 18% *(when 1 twin has schizophrenia, 18% of the time the other twin has schizophrenia)*
Genes and Individual Development
Sperm and Egg Formation

LO 2.6 Sperm and Egg Formation

• Gametes- sex cells. Only cells in humans that do not contain 46 chromosomes.

• These reproductive cells form in testes (in males) and ovaries (in females) through meiosis.
Sperm and Egg Formation

Cell Division

Figure 2.5  The Creation of Gametes Through Meiosis

How does meiosis differ from mitosis?

Miosis in males is complete before sperm are released.

- Miosis is complete in females when sperm fertilizes the egg.
Figure 2.6  Ovulation Process  The two ovaries alternate ovulation in each monthly cycle.
<table>
<thead>
<tr>
<th>Trimester</th>
<th>Period</th>
<th>Weeks</th>
<th>Milestones</th>
</tr>
</thead>
<tbody>
<tr>
<td>First</td>
<td>Germinal</td>
<td>1–2</td>
<td>Zygote divides and forms blastocyst, which implants in uterus and begins forming the amnion, placenta, and umbilical cord</td>
</tr>
<tr>
<td></td>
<td>Embryonic</td>
<td>3–4</td>
<td>Three layers form: the ectoderm, mesoderm, and endoderm; neural tube develops and produces neurons; heart begins beating; ribs, muscles, and digestive tract form</td>
</tr>
<tr>
<td></td>
<td></td>
<td>5–8</td>
<td>Arms and legs develop, then fingers and toes; placenta and umbilical cord function; digestive system develops; liver produces blood cells; embryo responds to touch</td>
</tr>
<tr>
<td>Fetal</td>
<td></td>
<td>9–12</td>
<td>Genitals form and release sex hormones; fingernails, toenails, and taste buds develop; heartbeat audible with stethoscope</td>
</tr>
<tr>
<td>Second</td>
<td></td>
<td>13–24</td>
<td>Mother feels movement; fetus kicks, turns, hiccups, sucks thumb, breathes amniotic fluid; responds to sounds, especially music and familiar voices; vernix and lanugo develop on skin</td>
</tr>
<tr>
<td>Third</td>
<td></td>
<td>25–38</td>
<td>Lungs develop fully; over two-thirds of birth weight is gained; brain development accelerates; sleep–wake cycles resemble newborn’s</td>
</tr>
</tbody>
</table>
Prenatal Care

Traditional Cultures and Beliefs

• Prenatal beliefs are impacted by generational wisdom of the time
• These beliefs can include
  ▪ Avoiding wine
  ▪ Specific types of meat
  ▪ Certain types of hot, cold food
  ▪ Witches
  ▪ Strong foods

LO 2.11 Prenatal Care in Traditional Cultures
Prenatal Care
Scientifically Based Prenatal Care

• Some current science-driven prenatal methods are derived from traditional cultures which include massages
• Current care can vary by ethnicity and SES
• Developing world less likely to receive prenatal care
• Current focus on diet, exercise, and teratogens

LO 2.12 Scientifically Based Prenatal Care
### TABLE 2.3 Essentials of Prenatal Care

#### Before Pregnancy

- Have a medical examination to ensure there are no diseases that may affect prenatal development. If not fully vaccinated, obtain vaccinations for diseases, such as rubella, that can damage prenatal development. (Vaccinations may be unsafe during pregnancy.)
- Avoid tobacco, alcohol, and other drugs, which may make it more difficult to become pregnant and are damaging to prenatal development.

#### During Pregnancy

- **Diet.** Maintain a balanced diet, including protein, grains, fruits, and vegetables. Avoid excessive fats and sugars and obtain sufficient iron and iodine. Gain 25–35 pounds in total; avoid dieting as well as excessive weight gain.
- **Exercise.** Engage in mild to moderate exercise regularly, including aerobic exercise, to stimulate circulatory system and muscles, as well as Kegel exercises to strengthen vaginal muscles. Avoid strenuous exercise and high-risk sports, such as long-distance running, contact sports, downhill skiing, waterskiing, and horseback riding.
- **Teratogens.** Avoid tobacco, alcohol, and other drugs. Avoid exposure to X-rays, hazardous chemicals, and infectious diseases.
- **Get regular prenatal checkups.** Regular medical visits are recommended during the entire pregnancy, beginning between week 8 and week 12. Ideally, women will see a doctor every four weeks after week 12, until week 28, when weekly visits are recommended for the duration of the pregnancy.
Prenatal Care

Diet

• Two key vitamins are iodine and iron
• Iodine deficiency tends to be a bigger issue in developing countries
• Iron deficiency puts women at risk for pre-term or low-birth-weight babies.
Prenatal Care

Exercise

• Continued physical activity is encouraged during pregnancy
• Aerobic exercise encourages good cardiovascular health and positively benefits the fetus
• A Non-aerobic exercise called Kegel exercises are also encouraged

LO 2.12 Scientifically Based Prenatal Care
Teratogens are behaviors, environments, and bodily conditions that could be harmful to the developing organism.

- Include tobacco, alcohol, prescription medications and other drugs

- The physical environment could also be a teratogen due to malnutrition or exposure to hazardous chemicals

LO 2.13 Teratogens
Teratogens
Timing of Teratogens

• Teratogens can impact the developing fetus and embryo at any time
• There does appear to be a critical period of prenatal development centered in the embryonic period
Figure 2.7 Timing of teratogens. Vulnerability to teratogens is greatest in the embryonic period. Source: Moore, 1974.
Teratogens

• Major teratogens exist in both developed and developing countries
  ▪ Malnutrition
  ▪ Infectious Disease
  ▪ Alcohol
  ▪ Tobacco

LO 2.13 Teratogens
IF YOU ARE PREGNANT, OR THINK YOU MIGHT BE PREGNANT, TELL THE X-RAY TECHNOLOGIST BEFORE HAVING AN X-RAY COMPLETED

Safety sign. Source: http://www.compliancesigns.com
Teratogens Malnutrition

• Malnutrition - mostly likely the most common teratogen worldwide.
• Rural nature of half the world’s population impacts nutrition (diet is affected by seasons)
  summer & fall = more food
  winter & spring = less food
• Folic acid is also important in preventing spina bifida & anacephaly.
• Eat food that have folic acid before pregnancy

LO 2.13 Teratogens
Teratogens Malnutrition

• Developed countries have food available, but may have malnutrition
• May be deficient in specific vitamins or an improper diet may lead to obesity
  ▪ Maternal obesity is linked to baby complications
• Prenatal nutrition can also cause prenatal problems

LO 2.13 Teratogens
• Rubella (German Measles)
  ▪ Exposure during embryonic stage can lead to heart abnormalities and mental retardation
  ▪ Exposure during the fetal stage can lead to hearing problems and low birth weight

• Vaccination can help, but Rubella remains widespread in less developed countries

LO 2.13 Teratogens
AIDS damages the immune system

Three strategies can help prevent transmission:

- Effective medicines
- Cesarean sections for AIDS-infected moms
- Infant formula in place of breast feeding

LO 2.13 Teratogens
Teratogens

Drugs-Alcohol

- Widespread damage in developed countries
- Fetal Alcohol Spectrum Disorder - set of problems that occur as a consequence of maternal alcohol use during pregnancy. 
  - facial deformities, heart problems, cognitive problems...
- Increased effects as child develops

LO 2.13 Teratogens
Teratogens

Drugs-Tobacco

• Maternal smoking increases risk of miscarriages, premature birth and low birth weight
• Infant effects include difficulty breathing and impaired heart functioning

LO 2.13 Teratogens
Teratogens
Drugs-Tobacco

• Childhood effects include poorer language skills, attention and memory problems and behavior problems
• Secondhand smoke by fathers is also detrimental

LO 2.13 Teratogens
Teratogens
Drugs

• Malnutrition and diseases are common in developing countries
• Alcohol and tobacco are common in developed countries
• Prescription drugs can be damaging
• Other teratogens include non-prescription drugs, severe stress, pollution and radiation

LO 2.13 Teratogens
Pregnancy Problems
Chromosomal Disorders

Chromosomal disorders can occur because of errors during meiosis

- This may cause too many or too few chromosomes in the cells of the zygote
- 45, 47, 48, 49 chromosomes

Two types of chromosomal disorders:

- Sex chromosome disorders
- Disorder on the 21st chromosome (Down Syndrome)
Sex Chromosome Disorders

Sex chromosomal disorders can result from extra X, an extra Y, or only an X and no second chromosome.

- Common consequences of sex chromosome disorders include:
  - Cognitive Deficit (mild-----severe)
  - Learning disorder
  - Speech impairments
  - Abnormality in development of reproductive system (becomes noticeable at puberty)
  - Underdeveloped testes, penis
  - No ovulation in girls
Examples

1. **XXY** = Klinefelter's syndrome
   weak muscles, low sperm count, delayed language

2. **X0** = Turner syndrome
   short, no menstrual cycle,

3. **XYY** = accelerated physical growth, some w/learning delays
Down Syndrome
Trisomy 21

• Distinct physical characteristics
• Cognitive Deficits
  ▪ Speech problems
  ▪ Mental retardation
• Problems in physical development
• Lower life expectancy

LO 2.14 Chromosomal Disorders
Chromosomal disorders are **NOT** passed from parent to child.

Relationship between maternal age and chromosomal disorders.

There may be a relation between a father’s age and chromosomal disorders, but it isn’t as clear.
Figure 2.8  Chromosomal Disorders and Maternal Age  Why does the risk rise so steeply after age 40? Source: Reproductive Medicine Associates of New Jersey, 2002
Prenatal Diagnosis

LO 2.15 Prenatal Diagnosis

• Ultrasounds-Uses high frequency sound waves
• Amniocentesis-Utilizes amniotic fluid
• Chorionic Villus Sampling-Utilizes cells from forming umbilical cord
Villus cells are collected via a needle...

Fluid is withdrawn and fetal cells are cultured.

Chorionic villi in placenta

Fetus (8–10 weeks)

Uterine cavity

Vagina

Ultrasound transducer

Needle

Placenta

Amniotic fluid

Uterus

Fetus (14–16 weeks)

or, a thin tube is inserted into the uterus to gather a sample from the placenta.
Infertility Causes

• For men- three main causes:
  ▪ Too few sperm
  ▪ Quality of sperm
  ▪ Low motility (movement)

• For women- many causes
  ▪ Most often caused by problems in ovulation

What are some causes of the inability to ovulate?

LO 2.16 Cause of Infertility
Question

What are the causes of infertility in men?

What are the causes of infertility in women?
Answer

**MEN**
- Disease/defects in sperm manufacturing process
- Genetics
- Drug abuse
- Alcohol abuse
- Cigarette smoking
- Age (+40 yrs) 3x longer to impregnate

**Women**
- Disease
- Drug abuse
- Alcohol abuse
- Cigarette smoking
- Being extremely overweight or underweight
Figure 2.9  Fertility and Maternal Age  Why does fertility decline after the mid-twenties?  
Source: Reproductive Medicine Associates of New Jersey, 2002
Infertility Treatments

• Historically, infertility was considered a female problem. Treatments included:
  - Giving more attention to wife to achieve mutual orgasm
  - Surgery on a woman’s anatomy
  - Bloodletting

LO 2.17 Infertility Treatments
Infertility Treatments

• Current treatments utilize Assisted Reproductive Technologies (ART)
  ▪ Artificial Insemination
    – Injects sperm into woman’s uterus
  ▪ Fertility Drugs
    – Mimic hormones involved in ovulation
  ▪ In-vitro fertilization
    – Ova removed and fertilized outside the womb then placed into uterus
Map 2.2  The “Infertility Belt”  In certain countries in Central Africa, infertility rates are as high as 30%. The reasons for this are unclear although malnutrition and high rates of STIs are probable factors.