Chapter 2

GENETICS AND PRENATAL DEVELOPMENT
Genetic Basics
Building Blocks of Life

- Chromosomes
- DNA
- Genes
- Genomes

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
  - What is seen or observed and can include a wide range of things

LO 2.1 Genotype and Phenotype
<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>Nearsighteded 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>
Genetic Basics
Expression of Traits

- Incomplete Dominance
  - Phenotype influenced primarily but, not exclusively, by dominant gene

- Polygenic Inheritance
  - Interaction of multiple genes

Figure 2.3  Incomplete Dominance in Sickle-Cell Inheritance
Two recessive genes for the sickle-cell trait results in sickle-cell anemia, but having one dominant and one recessive genes provides protection against malaria.

© 2013 by Pearson Education, Inc. All rights reserved.
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?
Genes and Individual Development
Sperm and Egg Formation
Cell Division

Figure 2.5  The Creation of Gametes Through Meiosis

How does meiosis differ from mitosis?

- Chromosome pairs split into single chromosomes
- Chromosomes replicate
- Crossing over: chromosomes swap sections of DNA
- Cell divides into two
- Pairs separate and cells divide again
Sperm and Egg Formation

LO 2.6 Sperm and Egg Formation

• Gametes- only cells in humans that do not contain 46 chromosomes.
• These reproductive cells form in testes (in males) and ovaries (in females) through meiosis.
The Beginning of Life
Conception

- Ovulation (release of ovum) occurs about 14 days into a woman’s cycle.
- For conception to occur, an ovum must be released and a sperm must travel up the fallopian tubes.

LO 2.7 Fertilization and Conception
The Germinal Period
First 2 Weeks of Life

• During the travel from the fallopian tube to the uterus cell division is taking place
  ▪ Blastocyst
  ▪ Trophoblast
  ▪ Embryonic disk

LO 2.8 Germinal Period
<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>
The Embryonic Period
Week 3-Week 8

Organs and structures are forming including

- Heartbeat
- Eyes, nose and mouth
- Bone development
- Arm and leg buds
- Digestive system

LO 2.9 Embryonic Period
The Fetal Period

Week 9-Birth

- The longest period of prenatal development
  - Heartbeat can be heard
  - Movement can be felt
  - Responds to sounds
  - Main impediment to viability is lung development

LO 2.10 Fetal Period
The Fetal Period
Week 9-Birth
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
### 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.

LO 2.12 Scientifically Based Prenatal Care
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
Prenatal Care
Teratogens

• Teratogens are environmental and bodily conditions that could be harmful
• 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

LO 2.13 Teratogens
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
Safety sign. Source: http://www.compliancesigns.com
AIDS

Three strategies can help prevent transmission:
- Effective medicines
- Cesarean sections for AIDS-infected moms
- Infant formula in place of breast feeding
Teratogens
Drugs-Alcohol

• Widespread damage in developed countries
• Fetal Alcohol Spectrum Disorder
• Increased effects as child develops
Teratogens
Drugs-Tobacco

• Maternal smoking increases risk of miscarriages, premature birth and low birth weight
• Infant effects include difficulty breathing and impaired heart functioning
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
Sex Chromosome Disorders
LO 2.14 Chromosomal 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
    - Abnormality in development of reproductive system (becomes noticeable at puberty)
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
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
Infertility Causes

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

• 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
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