Chapter 2
Genetics and Prenatal Development
Learning Objectives (1 of 4)

2.1 Distinguish between genotype and phenotype and identify the different forms of genetic inheritance.

2.2 Describe the sex chromosomes and identify what makes them different from other chromosomes.

2.3 Explain how behavior geneticists use heritability estimates and concordance rates in their research.

2.4 Describe how the concept of epigenesis frames gene–environment interactions, and connect epigenesis to the concept of reaction range.
2.5 Explain how the theory of genotype environment effects casts new light on the old nature–nurture debate.

2.6 Outline the process of meiosis in the formation of reproductive cells.

2.7 Describe the process of fertilization and conception.

2.8 List the major causes and treatments for infertility, and describe how infertility is viewed in different cultures.
Learning Objectives (3 of 4)

2.9 Describe the structures that form during the germinal period.

2.10 Outline the major milestones of the embryonic period.

2.11 Describe the major milestones of the fetal period and identify when viability occurs.

2.12 Compare and contrast prenatal care in traditional cultures and developed countries.

2.13 Identify the major teratogens in developing countries and developed countries.
2.14 Explain how chromosomal disorders occur.

2.15 Describe causes and symptoms of some common genetic disorders.

2.16 Describe the three main techniques of prenatal diagnosis.

2.17 Explain who is likely to seek genetic counseling and for what purpose.
Genetic Basics: The Sex Chromosomes

• The 23\textsuperscript{rd} pair of chromosomes determines male or female

• Males more vulnerable to X-linked recessive disorders

• X-linked inheritance
Prenatal Care (1 of 2)

• Prenatal beliefs are impacted by generational wisdom of the time

• These beliefs can include
  – Avoiding wine (Beng)
  – Specific types of meat (Beng)
  – Certain types of hot or cold food (Bali)
  – Witches (Bali)
Prenatal Care: (2 of 2)

• Developing countries have developed specific beliefs as well

• These scientific beliefs include
  – Women typically gain 25-35 pounds
  – Women should receive regular evaluations

• Current care can vary by ethnicity and SES

• Developing world less likely to receive prenatal care
Map 2.1 Ethnic Variations in Prenatal Care within the United States

How does prenatal care differ for White women compared with other ethnic groups? What economic factors might account for these variations?
Teratogens (1 of 3)

- Teratogens are environmental and bodily conditions that could be harmful
- They can include tobacco, alcohol, prescription medications, and other drugs
- The physical environment could also be a teratogen due to malnutrition or exposure to hazardous chemicals
Malnutrition is the most common worldwide teratogen

Eating healthy diets is recommended but not likely in developing countries

Living in rural areas could also impact prenatal health

Folic acid can reduce incidences of anencephaly and spina bifida
Teratogens (3 of 3)

- Two key vitamins are iodine and iron
- Iodine deficiency tends to be a bigger issue in developing countries
- Iron is necessary for a healthy pregnancy
Vulnerability to teratogens is greatest in the embryonic period.
Teratogens: Infectious Diseases (1 of 2)

- 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
AIDS (Acquired Immune Deficiency Syndrome)

Three strategies can help prevent transmission

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

- Widespread damage in developed countries
- Fetal alcohol spectrum disorder
  - Includes facial deformities, heart problems, misshapen limbs, and a variety of cognitive problems
  - Increased effects as child develops
Teratogens: Nicotine

- Maternal smoking increases risk of miscarriages, premature birth, and low birth weight
- Infant effects include difficulty breathing and impaired heart functioning
- Childhood effects include poorer language skills, attention and memory problems, and behavior problems
- Secondhand smoke by fathers is also detrimental
Other Teratogens

- Alcohol and tobacco are common in developed countries

- Recreational psychoactive drugs cause physical, cognitive and behavioral problems
  - Marijuana affects development of the brain and CNS
  - Cocaine can result in low birth weight
  - Heroin can lead to painful withdrawal

- Prescription drugs can be damaging
Teratogens

Stress

- When you’re stressed, your body goes into "fight or flight" mode, sending out a burst of cortisol and other stress hormones. These are the same hormones that surge when you are in danger. They prepare you to run by sending a blast of fuel to your muscles and making your heart pump faster.

- When the stress goes away the body and brain goes back to its normal state and is balanced once more.

- Chronic stress, stress that won’t let up may also contribute to subtle differences in brain development that might lead to behavioral issues as the baby grows, she adds.
Pregnancy Problems
Chromosomal Disorders

• Chromosomal disorders can occur because of an issue during meiosis
  – This may cause too many or too few chromosomes in the cells of the zygote

• Two types of chromosomal disorders are:
  – Sex chromosome disorders
  – Disorder on the 21st chromosome (Down syndrome)
Sex Chromosome Disorders

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

- **Klinefelter syndrome**

- **Turner syndrome**
  - Common consequences of sex chromosome disorders include:
    - Cognitive deficit
    - Abnormality in reproductive system at puberty
Down Syndrome Trisomy-21

- Identifiable by physical characteristics
- Cognitive deficits
  - Speech problems
  - Mental retardation
- Social development varies
- Lower life expectancy
Parental Age and Chromosomal Disorder

- Chromosomal disorders tend to NOT be passed from parent to child
- Relationship between maternal age and chromosomal disorders
- There may be a relationship between a father’s age and chromosomal disorders but it isn’t as clear
Figure 2.9 Down Syndrome and Maternal Age

Why does the risk rise so steeply after age 40?
Prenatal Diagnosis

- Ultrasounds – uses high frequency sound waves

- Amniocentesis – utilizes amniotic fluid
  - 15 to 20 weeks into pregnancy

- Chorionic villus sampling – utilizes cells from forming umbilical cord
  - 5 to 10 weeks into pregnancy
Genetic Counseling

- Before having children some may seek out genetic counseling
  - Inherited genetic condition
  - Couples with history of miscarriage or infertility
  - Older couples (women over 35, men over 40)
Learning Objectives (1 of 4)

3.1 Describe the three stages of the birth process.

3.2 Name two common types of birth complications and explain how they can be overcome by cesarean delivery.

3.3 Summarize the history of birth in the West from the 15th century to today.

3.4 Describe cultural variations in birth beliefs and identify who may assist with the birth.
3.5 Compare and contrast cultural practices and medical methods for easing the birth process.

3.6 Identify the features of the two major scales most often used to assess neonatal health.

3.7 Identify the neonatal classifications for low birth weight and describe the consequences and major treatments.

3.8 Describe the differences in maternal and neonatal mortality both within and between developed countries and developing countries.
Learning Objectives (3 of 4)

3.9 Describe neonates’ patterns of waking and sleeping, including how and why these patterns differ across cultures.

3.10 Describe the neonatal reflexes, including those that have a functional purpose and those that do not.

3.11 Describe the neonate’s sensory abilities with respect to touch, taste and smell, hearing, and sight.

3.12 Describe the cultural customs surrounding breast feeding across cultures and history.
Learning Objectives (4 of 4)

3.13 Identify the advantages of breast feeding and where those advantages are largest.

3.14 Describe neonates’ types of crying and how soothing methods vary across cultures.

3.15 Describe the extent to which human mothers “bond” with their neonates and the extent to which this claim has been exaggerated.

3.16 Describe the reasons for postpartum depression and its consequences for children.
Birth and Its Cultural Context
Figure 3.1 The Three Stages of the Birth Process

Stage 1: Labor

- Umbilical cord
- Placenta
- Cervix

Contractions increase in duration, frequency, and intensity, causing the cervix to dilate.

Stage 2: Delivery

- Uterus
- Placenta

The mother pushes, and the baby crowns and then exits the birth canal and enters the world.

Stage 3: Expelling of Placenta & Umbilical Cord

- Placenta
- Umbilical cord

Contractions continue as the placenta and umbilical cord are expelled.

Which stage is longest and most difficult?
Stages of the Birth Process: The First Stage: Labor

- Longest and most taxing stage
- Contractions in the uterus cause cervix to dilate
- Labor is painful but there are some ways to ease discomfort
  - Rocking chair, warm shower or bath, massages, or taking a walk
  - Emotional support is important
Stages of the Birth Process: The Second & Third Stage

- Delivery can take up to an hour
- Crowning occurs
- May be given an episiotomy
- Third stage expels the placenta and umbilical cord
- Complications can occur if placenta not fully expelled
Two common birth complications:

- Failure to progress
- Breech presentation

Cesarean delivery can be done to deal with birth complications
Stages of the Birth Process: Birth Complications (2 of 2)

- Cesarean – retrieving the baby directly from the uterus
- High rates of C-section may be due to extreme caution
- Vaginal birth after C-section is possible
Map 3.1 Cesarean Section Rates, Selected Countries

Which countries have the highest rates of cesarean sections? What determines whether a country has high or low rates?
Peculiar History of Birth in the West (1 of 2)

• Pre-15th century – midwives respected

• 15th century – midwives suspected of being witches

• 18th century – medical schools and physician delivery
  – Field of obstetrics
20th century – doctors not properly trained

- Led to misuse of drugs for delivery including morphine
- Late 20th century backlash led to advocating natural childbirth

With hospital deliveries, no difference in maternal health or neonatal outcomes between natural childbirth and medical methods
Cultural Variations in Birth Beliefs and Practices (1 of 3)

• Celebration of birth
  – ILA of Zimbabwe praise the woman and offer gifts

• Fear or wariness of birth
  – Arapesh of New Guinea allow birth on outskirts of the village in a place reserved for menstruation activities
Cultural Variations in Birth Beliefs and Practices (2 of 3)

- Cultures may have purifying traditions for mothers after birth
- Placenta also has meanings
  - Some cultures bury in sacred place
  - Some cultures believe it has value for hormones and nutrients
Cultural Variations in Birth Beliefs and Practices (3 of 3)

• Midwives maintain birth assistant in traditional cultures
• Variation in how one becomes a midwife
• Some variation in how midwives are viewed
Cultural Variations in Methods for Easing the Birth (1 of 2)

• Attempts to ease birth process include
  – Herbal medicines to cope with pain
  – In the Ukraine, traditionally a woman may be given a glass of whiskey
  – Midwives give instruction and encouragement as labor progresses
  – In some traditional cultures, midwife may call on spiritual assistance
Cultural Variations in Methods for Easing the Birth (2 of 2)

- Emotional and social support important

- Medical use of epidural seen in developed countries

- Birthing position also eases pain
  - Upright, semi-sitting, half reclining position

- Placenta delivered through various methods

- Umbilical cord may be cut and tied
The Neonate
The Neonate at Birth

• Neonate:
  – Fuzzy hair called lanugo
  – Misshapen head with soft spots called fontanels
  – About 20 inches and 7.5 pounds
Half of all neonates will have neonatal jaundice

Effective treatment is phototherapy

First few minutes after birth are critical
  - Anoxia is a risk
Measuring Neonatal Health (2 of 4)

- Neonatal assessment is critical in first few minutes
- Apgar Scale
  - Assessed on Appearance, Pulse, Grimace, Activity, and Respiration
  - Rated on each category with a 0 to 2 score
  - Gives total score of 0 to 10
• Measured twice, in first minute and after five minutes

• Brazelton Neonatal Behavioral Assessment Scale (NBAS)
  – Rates neonates on 27 items
  – Receives rating of worrisome, normal, or superior
  – Most effective if given at one day and a week later
Measuring Neonatal Health (4 of 4)

- Can help parents interact with infants
- Useful in research on cultural differences in parenting practices
**Table 3.1 The Apgar Scale**  
Total Score: 7–10 = Good to excellent condition; 4–6 = Requires assistance to breathe; 3 or below = Life-threatening danger

<table>
<thead>
<tr>
<th>Subtest</th>
<th>0</th>
<th>1</th>
<th>2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Appearance (Body color)</td>
<td>Blue and pale</td>
<td>Body pink, but extremities blue</td>
<td>Entire body pink</td>
</tr>
<tr>
<td>Pulse (Heart rate)</td>
<td>Absent</td>
<td>Slow—less than 100 beats per minute</td>
<td>Fast—100 to 140 beats per minute</td>
</tr>
<tr>
<td>Grimace (Reflex irritability)</td>
<td>No response</td>
<td>Grimace</td>
<td>Coughing, sneezing, and crying</td>
</tr>
<tr>
<td>Activity (Muscle tone)</td>
<td>Limp and flaccid</td>
<td>Weak, inactive, but some flexion of extremities</td>
<td>Strong, active motion</td>
</tr>
<tr>
<td>Respiration (Breathing)</td>
<td>No breathing for more than 1 minute</td>
<td>Irregular and slow</td>
<td>Good breathing with normal crying</td>
</tr>
</tbody>
</table>
The Neonate’s Health (1 of 3)

- Low birth weight (LBW) – less than 5.5 pounds (2,500 grams)
  - Pre-term
  - Small for date

- Very low birth weight – less than 3.3 pounds (1,500 grams)

- Extremely low birth weight – less than 2.2 pounds (1,000 grams)
The Neonate’s Health (2 of 3)

• Causes for LBW

• Developing countries – malnourished mothers and lack of prenatal care

• Developing countries – cigarette smoking, alcohol and drug use, maternal age (younger than 17 or older than 40), multiple births
The Neonate’s Health (3 of 3)

- Consequences of LBW
  - High mortality rate
  - Inadequately developed physical systems
  - Immature lungs, immune system, and CNS
  - Problems long term include risk of asthma, language delays, poor school performance, attention deficits, and low educational attainment
Map 3.2 Rates of Low Birth Weight Around the World

Why are rates so high in developing countries?
Treatments for LBW infants include

- Kangaroo care: skin to skin contact for 2 to 3 hours a day

- Infant massages
  - Triggers hormonal release that promotes weight gain, muscle development, and neurological development
Neonatal and Maternal Mortality

- Maternal mortality has declined in developing countries over the past 30 years.
- Within developing countries, neonatal and maternal mortality is varied.
- Neonatal mortality rate variation tends to be a function of poverty and access to high quality medical care.
- Maternal mortality rates are rising in certain groups.
Neonatal Sleeping Patterns

- Neonates sleep/wake cycle governed more by hunger than day/night cycle
- REM sleep dominates
- For neonates REM sleep stimulates brain activity
- Traditional cultures keep close physical contact
  - May cause infants to spend more day hours sleeping than in industrialized cultures
### Table 3.2 Neonatal Reflexes

<table>
<thead>
<tr>
<th>Reflex</th>
<th>Stimulation</th>
<th>Response</th>
<th>Disappears by</th>
</tr>
</thead>
<tbody>
<tr>
<td>Stepping</td>
<td>Hold baby under arms with feet touching floor</td>
<td>Makes stepping motions</td>
<td>2 months</td>
</tr>
<tr>
<td>Moro</td>
<td>Dip downward suddenly, or loud sound</td>
<td>Arch back, extend arms and legs outward, bring arms together swiftly</td>
<td>3 months</td>
</tr>
<tr>
<td>Babkin</td>
<td>Press and stroke both palms</td>
<td>Mouth opens, eyes close, head tilts forward</td>
<td>3 months</td>
</tr>
<tr>
<td>Sucking</td>
<td>Object or substance in mouth</td>
<td>Sucking</td>
<td>4 months</td>
</tr>
<tr>
<td>Rooting</td>
<td>Touch on cheek or mouth</td>
<td>Turn toward touch</td>
<td>4 months</td>
</tr>
<tr>
<td>Grasping</td>
<td>Object placed in palm</td>
<td>Hold tightly</td>
<td>4 months</td>
</tr>
<tr>
<td>Swimming</td>
<td>Baby is immersed in water</td>
<td>Holds breath, swims with arms and legs</td>
<td>4 months</td>
</tr>
<tr>
<td>Babinski</td>
<td>Stroke sole of foot</td>
<td>Foot twists in, toes fan out</td>
<td>8 months</td>
</tr>
</tbody>
</table>
Neonatal Senses (1 of 2)

- Touch – earliest sense to develop
  - Neonates do experience pain
- Taste and smell – well developed
  - Show smell preferences for mother’s diet
  - Prefer sweet taste
  - Prefer mother’s breast smell to another’s
Neonatal Senses (2 of 2)

• Hearing – well developed
  – Sensitive to human speech
  – Prefer mother’s voice
  – Have problems with sound localization

• Sight – least developed
  – Sees 8 to 14 inches
  – Binocular and color vision limited
  – Prefer patterns to random designs
  – Prefer faces to other patterns
Caring for the Neonate
Perspectives on Breast Feeding (1 of 2)

• Historical evidence
  – Mother and infant reflex actions at birth
    ▪ Let-down reflex
  – Historical evidence suggests breast feeding common
  – Substitute milk, wet nurse, formula became alternatives
Breast feeding back on the rise

- Mother’s age, education, and SES increase likelihood of breast feeding
Benefits of Breast Feeding (1 of 2)

• Benefits include
  – Colostrum
  – Disease protection
  – Cognitive development
  – Reduced obesity
  – Better health in childhood and adulthood

• Mothers benefit by reduced bleeding, strengthening bones, and reducing risk of ovarian cancer
Benefits of Breast Feeding (2 of 2)

• Benefits of breast feeding extremely important in developing countries
  – More illness in developing countries
  – Not receiving vaccinations

• Issues with breast feeding in developing countries
  – May have infectious disease (maternal)
  – Using formula mixed with non purified water could contribute to death rates
Map 3.3 Percentage of Infants Exclusively Breast-Fed at 4 Months of Age
In their first months of life, infants often cry for no apparent reason.
Table 3.3 Period of PURPLE Crying in the Early Months

<table>
<thead>
<tr>
<th></th>
<th>Peak Pattern</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>P</td>
<td>Peak Pattern</td>
<td>Crying peaks around age 2 months and then declines</td>
</tr>
<tr>
<td>U</td>
<td>Unpredictable</td>
<td>Crying in the early months often comes and goes unpredictably, for no apparent reason</td>
</tr>
<tr>
<td>R</td>
<td>Resistant to soothing</td>
<td>Crying may continue despite parent’s best soothing efforts</td>
</tr>
<tr>
<td>P</td>
<td>Pain-like face</td>
<td>Crying babies may look like they are in pain even though they are not</td>
</tr>
<tr>
<td>L</td>
<td>Long lasting</td>
<td>Babies cry for longer in the early months, sometimes 30-40 minutes or more</td>
</tr>
<tr>
<td>E</td>
<td>Evening crying</td>
<td>Babies usually cry most in the afternoon and evening</td>
</tr>
</tbody>
</table>
Crying and Soothing

- Duration and intensity of crying differ between cultures
- Episodes are longer and more intense in cultures where infants are left on their own a lot.
- Traditional cultures typically hold neonate for most of the day
- Swaddling babies has been shown to reduce crying
  - 10% of Western babies are colicky with no known cause
- Soothing a crying neonate includes
  - Lifting baby by shoulder, soothing repetitive movements, soothing sounds, and distraction
- Common theme is new source of stimulation
Bonding: Myth and Truth

• No support that first hour is critical for bonding in mother and infant

• Hospitals still encourage close contact immediately after birth
Postpartum Depression

• In some cases birth can cause postpartum depression

• Combination of hormonal changes and deep feelings of anxiety, sadness, and difficulty sleeping

• Increased risk if previous episodes of depression

• Some have genetic vulnerability for depression

• Lack of social support

• May impact child development