Chapter 2

GENETICS AND PRENATAL DEVELOPMENT

Instructor: Monica Moreno
Tuesday evening class: 6:50 pm to 10:00 pm
Section: 3080
Fall 2015
Learning Objectives

- **LO 2.1** Distinguish between genotype and phenotype and identify the different forms of genetic inheritance
- **LO 2.2** Describe the sex chromosomes and identify what makes them different from other chromosomes
- **LO 2.3** Describe how behavior geneticists use heritability estimates and concordance rates in their research
- **LO 2.4** Describe how the concept of epigenesis frames gene–environment interactions, and connect epigenesis to the concept of reaction range
- **LO 2.5** Explain how the theory of genotype → environment effects casts new light on the old nature–nurture debate
- **LO 2.6** Outline the process of meiosis in the formation of reproductive cells and specify how the process differs for males and females
- **LO 2.7** Describe the process of fertilization and conception
- **LO 2.8** Describe the structures that form during the germinal period, and identify when implantation takes place.
Learning Objectives

• LO 2.9 Outline the major milestones of the embryonic period and identify when they take place
• LO 2.10 Describe the major milestones of the fetal period and identify when viability occur
• LO 2.11 Recall some approaches to prenatal care in traditional cultures
• LO 2.12 Summarize scientifically based information on prenatal care
• LO 2.13 Identify the major teratogens in developing countries and developed countries
• LO 2.14 Explain how chromosomal disorders occur
• LO 2.15 Describe the three main techniques of prenatal diagnosis, and explain who is likely to seek genetic counseling and for what purposes
• LO 2.16 List the major causes of infertility for both men and women
• LO 2.17 Describe the current treatments for infertility
• LO 2.18 Compare rates of infertility worldwide, and contrast the views of infertility in developed and developing countries
Prenatal beliefs are impacted by generational wisdom of the time. These beliefs can include:

- **Avoiding wine**: West Nigerian African women are advised to avoid palm wine while pregnant.
- **Specific types of meat**: Also not to eat bushbuck antelope because the baby will be born with strips.
- **Certain types of hot, cold food**: In Indonesia pregnant women are told to avoid different types of hot or cold food.
- **Witches**: Are a common belief in both cultures although in different ways. In Africa if a pregnant mother cast a curse on someone the baby will become a witch. In Indonesia a witch is very attracted to the blood of a pregnant woman therefore they wear a protective charm.
- **Strong foods**: In Australia, the Aboriginal culture believe women should avoid from eating strong foods like emu, rabbit and bandicoot. It could make the pregnancy and birth dangerous to the mother.
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.
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?
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
• 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
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
Malnutrition- mostly likely the most common teratogen worldwide.

Rural nature of half the world’s population impacts nutrition (diet is affected by seasons)
Teratogens
Malnutrition

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

Infectious Diseases

• AIDS

• 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
- 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 can occur because of errors during meiosis:
- This may cause too many or too few chromosomes in the cells of the zygote.

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
• **Klinefelter syndrome**
• **Turner syndrome**
  ▪ 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
Parental Age and Chromosomal Disorder

- 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

LO 2.14 Chromosomal Disorders
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
Infertility Causes

• For men- three main causes:
  ▪ Too few sperm- Low sperm count
  ▪ Quality of sperm- Sperm is not viable
  ▪ Low motility-movement of the sperm

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

What are some causes of the inability to ovulate?
  ▪ Age
  ▪ Disease
  ▪ Drug/alcohol abuse
  ▪ Cigarette smoking

LO 2.16 Cause of Infertility
Figure 2.9  Fertility and Maternal Age  Why does fertility decline after the mid-twenties?  
Source: Reproductive Medicine Associates of New Jersey, 2002
Historically, infertility was considered a female problem. Treatments included:

- Giving more attention to wife to achieve mutual orgasm - It was believed in the 1800’s that a seed was needed and released from the man and a woman when they would orgasm.
- Surgery on a woman’s anatomy
- Bloodletting - Which means a doctor would cut a vessel and let the women bleed out.

Only in the last 50 years is it known that in men and women contribute equally to infertility.
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.
Birth and Its Cultural Context
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

LO 3.1 Stages of birth
Figure 3.1  The Three Stages of the Birth Process  Which stage is longest and most difficult?
Stages of the Birth Process: The Second Stage

• Delivery can take up to an hour, but there is wide variation
• Crowning - baby’s head appears at outer opening of vagina.

LO 3.1 Stages of birth
Stages of the Birth Process: The Third Stage

- placenta and umbilical cord are expelled
- Complications can occur if placenta is not fully expelled
Stages of the Birth Process

Birth Complications

Two common birth complications:
• Failure to progress
• Breech Presentation

Cesarean delivery can be done to deal with birth complications

LO 3.2 Birth Complications
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?
Stages of the Birth Process

Birth Complications

• Cesarean-surgical incision to remove baby from the uterus
• High rates of C-section may be due to extreme caution
• Vaginal birth after C-section (VBAC) is possible
Cultural and Historical Variations in Birth Beliefs

- Celebration of birth
  - ILA of Zimbabwe praise the woman and offer gifts
- Fear or wariness of birth
  - Arapesh of New Guinea allow birth only on outskirts of village in a place reserved for menstruation & excretion activities.
Cultural and Historical Variations in Birth Beliefs

• 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

LO 3.3 Birth beliefs
Cultural Variations in Birth Beliefs
Midwives and Others

- Midwives maintain birth assistant in traditional cultures
- Variation in how one becomes a midwife
- Variation in how midwives are viewed

LO 3.4 Cultural practices in easing birth
Cultural Variations in Birth Practices

- Attempts to ease birth process include:
  - Abdominal massage and herbal teas
  - Herbal medicines to cope with pain
  - Midwives give instruction and encouragement
  - Symbols placed in different areas

LO 3.4 Cultural practices in easing birth
Cultural Variations in Birth Beliefs
Easing the Birth

• 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

LO 3.4 Cultural practices in easing birth
Historical Variations in Birth Beliefs
Peculiar History of Birth in the West

- Pre-15\textsuperscript{th} Century-Midwives respected
- 15\textsuperscript{th} Century-Midwives suspected of being witches
- 18\textsuperscript{th} Century-Medical schools and physician delivery

LO 3.5 History of birth in the West
Historical Variations in Birth Beliefs
Peculiar History of Birth in the West

- **20th Century** - Doctors not properly trained
  - Led to misuse of drugs for delivery including morphine
  - Late 20th century backlash led to advocating natural childbirth

LO 3.5 History of birth in the West
Historical Variations in Birth Beliefs

Peculiar History of Birth in the West

• Currently there are several improvements in birth process
  ▪ Collaboration during birth
  ▪ Fathers more likely involved
  ▪ Medications are safer
  ▪ Use of electronic fetal monitoring

LO 3.5 History of birth in the West
Cultural Variations in Neonatal & Maternal Mortality

- Maternal mortality has decreased in developing countries due to:
  - Better nutrition
  - Greater access to health care

- Rates of infant and maternal mortality are still higher in developing countries than in developed countries.

LO 3.6 Maternal and neonatal mortality
Cultural Variations in Neonatal & Maternal Mortality

• Substantial variation in neonatal and maternal mortality within developed countries.
  ▪ Neonatal mortality is twice as high for African Americans as for Whites.
  ▪ Rates of infant mortality among Latinos and Asian American are similar to those of Whites.

• Maternal mortality is over 3X as high among African Americans as among Whites.
  ▪ What are possible explanations?

LO 3.6 Maternal and neonatal mortality
How do neonatal and maternal mortality rates compare? What factors might explain why mortality rates are higher in developing countries than in developed countries? (continued on next slide)
Map 3.2  Neonatal and Maternal Mortality Worldwide  How do neonatal and maternal mortality rates compare? What factors might explain why mortality rates are higher in developing countries than in developed countries? (continued from previous slide)
The Neonate
The Neonate

LO 3.7 Assessing neonatal health

- Neonate:
  - Fuzzy hair called lanugo
  - Misshapen head with soft spots called fontanels
  - About 20 inches and 7.5 pounds
  - About half have jaundice
The Neonate’s Health
Measuring Neonatal Health

• Neonatal assessment is critical in first few minutes

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

LO 3.7 Scales to assess neonatal health
# 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>Score</th>
<th>0</th>
<th>1</th>
<th>2</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Appearance (Body color)</strong></td>
<td>Blue and pale</td>
<td>Body pink, but extremities blue</td>
<td>Entire body pink</td>
</tr>
<tr>
<td><strong>Pulse (Heart rate)</strong></td>
<td>Absent</td>
<td>Slow—less than 100 beats per minute</td>
<td>Fast—100–140 beats per minute</td>
</tr>
<tr>
<td><strong>Grimace (Reflex irritability)</strong></td>
<td>No response</td>
<td>Grimace</td>
<td>Coughing, sneezing, and crying</td>
</tr>
<tr>
<td><strong>Activity (Muscle tone)</strong></td>
<td>Limp and flaccid</td>
<td>Weak, inactive, but some flexion of extremities</td>
<td>Strong, active motion</td>
</tr>
<tr>
<td><strong>Respiration (Breathing)</strong></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>

**Source:** Based on Apgar, 1953
The Neonate’s Health
Measuring Neonatal Health

• Brazelton Neonatal Behavioral Assessment Scale (NBAS)
  ▪ Rates neonates on 27 items
  ▪ Rated as worrisome, normal or superior
  ▪ Most effective if given at one day and a week later
  ▪ Can help parents interact with infants
  ▪ Useful in research on cultural differences in parenting practices

LO 3.7 Assess neonatal health
Low Birth Weight

- Low birth weight (LBW)-less than 2500 grams
- Very Low birth weight-less than 1500 grams
- Extremely Low birth weight-less than 1000 grams
Main Causes for LBW

- Developing countries - malnourished mothers and lack of prenatal care
- Developed countries - cigarette smoking
- Other factors - multiple births, maternal age, drug use
Map 3.3 Rates of Low Birth Weight Around the World  Why are rates so high in developing countries?
Consequences of LBW

- High mortality rate
- LBW for small for date infants
  - Poor maternal malnutrition, illness, teratogen exposure
- LBW for pre-term neonates
  - Inadequately developed physical systems
  - Immature lungs, immune system, and CNS