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
Genetic Basics
Building Blocks of Life

- Chromosomes- There are 23 pairs of chromosomes with a total of 46.
- DNA- The chromosomes are composed of complex molecules called DNA.
- Genes- The DNA in the chromosome is organized into segments call GENES. These genes include hereditary information.
- Genomes- There are 23,000 genes in our 46 chromosomes which makes the total human genome.

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
Genetic Basics
Expression of Traits

• Dominant Genes-Is a gene that is dominant.

• Recessive Genes-Is a gene that is not common. It’s only evident when matched with another recessive gene.

• Allele-Several forms of a gene, usually arising through mutation, that are responsible for hereditary variation.

• Dominant-Recessive Inheritance- The dominant gene inherited by your parents influences the phenotype.

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>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>

Table 2.1  Traits With Single-Gene Dominant–Recessive Inheritance
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.
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?
• Behavior Genetics
  ▪ Estimating influence of genes and environment on development
  ▪ Studying twins is helpful to the study and helps understand the importance of genetics and consider how the environment effects behavior

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

• Concordance Rates
  ▪ Percentage that indicates degree of similarity in phenotype among pairs of family members

LO 2.3 Behavior Genetics
Genes and Individual Development
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.
The Germinal Period
First 2 Weeks of Life

During the travel from the fallopian tube to the uterus cell division is taking place

- **Blastocyst** - Ball of about 100 cells formed by about one week following conception. It implants itself to the uterine wall. The blastocyst has two layers the trophoblast.
- **Trophoblast** - Provides protection and nourishment.
- **Embryonic disk** - Is the inner layers of the cell which will become the embryo.
- **Placenta** - Is between the uterine wall and the disk.
- **Umbilical cord** - Is the cord that connects the embryo to the placenta. This is how the embryo receives nutrients.

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>9–12</td>
<td></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>13–24</td>
<td></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>25–38</td>
<td></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

- **Key Developments of the Embryonic Layers**
  - Ectoderm- Is the outer layer of the disk that forms. It will become the skin, hair, nails and sensory organs as well as the nervous system.
  - Mesoderm- Is the middle layer which will become the muscles, bones, reproductive system and circulatory system.
  - Endoderm- Is the inner layer and will become the digestive system and respiratory system.

LO 2.9 Embryonic Period
The Embryonic Period
Week 3-Week 8

Organs and structures are forming including:
- Heartbeat- In the forth week.
- Eyes, nose and mouth- In the forth week.
- Bone development- Begin to develop in the forth week.
- Arm and leg buds- 5 to 8 weeks begin to develop.
- Digestive system- 5 to 8 weeks begin to develop.
The Fetal Period
Week 9-Birth

• The longest period of prenatal development
  ▪ Heartbeat can be heard – During the 12th week
  ▪ Movement can be felt- By the 4th month.
  ▪ Responds to sounds- By the end of the 3rd trimester.
  ▪ Main impediment to viability is lung development- Develop toward the end of the pregnancy.

LO 2.10 Fetal Period
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 casts 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 eating strong foods like emu, rabbit and bandicoot. It could make the pregnancy and birth dangerous to the mother.

LO 2.11 Prenatal Care in Traditional Cultures
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.
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

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)

LO 2.13 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

LO 2.13 Teratogens
Teratogens
Infectious Diseases

• 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

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

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

LO 2.14 Chromosomal 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
Infertility Treatments

• 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.
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