Genes and Prenatal Development
Chromosomes and Genes

The Beginning of Life
Every cell in our body promotes growth and sustains life according to chemical instructions in their DNA.

Each molecule of our DNA is called a chromosome.

Humans have 46 chromosomes arranged in 23 pairs, with one exception.
Reproduction Cell

- The exception is the reproductive cell called a gamete. (sperm/ovum).

- When sperm and ovum combine they created a new cell called a zygote.

- The zygote is made 23 chromosomes from the mother and 23 chromosomes from the father. (all DNA instructions are in the zygote)
DNA instructions are organized into units called genes.

Each gene is located on a particular one of the 46 chromosomes.

Genes are passed down from generation to generation.
Genetic Variations

- Half a person’s genes come from each parent.
- ¼ from each grandparent
  - Maternal grandmother
  - Maternal grandfather
  - Paternal grandmother
  - Paternal grandfather
- 1/8 from each of our 8 great-grandparents.
Genetic Variation

- Together all the genes make up our genetic inheritance or genotype.
  - Genetic potential

- Phenotype: observable characteristics of person, including appearance, personality, intelligence and other traits.
Many genes are identical for every human being.

Some genes vary slightly based on their code.

Allele: any possible forms in which a gene for a particular trait can occur.
Sibling Similarities

- Full siblings get half their genes from each parent.
  - Which half they receive depends on which of the millions of possible gametes combined to start their development.
  - Every zygote is genetically unique.
Twins

- Exception to a zygote’s genetic unique.
- 1 out of every 250 conceptions result in twins.
  - Their zygote splits completely apart, creating two, four or eight separate zygotes.
    - If each of those implants and grows, multiple births.
Monozygotic twins (identical): originate from one zygote that splits early in development.

Because they originate from the same zygote, they have identical genetics instructions.

Dizygotic twins (fraternal): 1 in 125 conceptions, develop two zygotes created by two ova fertilized by two sperm (at the same time)
Twins

- Incidence of Dizygotic twins is genetic.
  - Women inherit the tendency to release more than one ovum when they ovulate.
    - Age and gender impact incidence.

- Incidence of monozygotic twins is not genetic.
In 22 of the 23 pairs of human chromosomes that each person inherits, the chromosomes are closely matched.

- The 23\textsuperscript{rd} pair of chromosomes is special
  - Females: XX (two X chromosomes)
  - Males: XY (one X and one Y)

Every egg in a female contains either one X or the other X.

In males, half of his sperm carry an X Chromosome and half carry a Y chromosomes.
  - *XY chromosomes create a boy.
  - *XX chromosomes create a girl.
Almost every trait is:

- Polygenic: meaning it is affected by many genes
- Multifactorial: meaning influenced by many factors.

- Additive heredity: a gene that adds up to some aspect of the phenotype.
  - Depends on additions from other genes.
Dominant-Recessive Heredity:

- Alleles interact and one allele is dominant and one is recessive.
  - Dominant gene is more influential
  - Recessive gene is more influential

Carriers: a person whose genotype includes a gene that is not expressed in their phenotype.
Genetic Diseases

- Alzheimer’s Disease
- Cancer
- Cleft Palate
- Diabetes
- Muscular Dystrophy
- Rett Syndrome
- Schizophrenia
- Tourette Syndrome
Three Stages/Periods of Prenatal Development

1. Germinal Period: Zygote
   - First 2 weeks
   - 60% of all developing organisms fail to implant

2. Embryonic Period: Embryo
   - 3rd week through the 8th week
   - 20% of all embryos are spontaneously aborted

3. Fetal Period: Fetus
   - 9th week through birth
   - 5% of all fetuses are aborted spontaneously aborted or stillborn

31% of all zygotes grow and survive to become a newborn babies!!
Pregnancy begins with conception.

However, pregnancy doesn’t affect the woman and can’t be confirmed by blood or urine tests until implantation.

Implantation is approximately 10 days after conception.
Length of Pregnancy

- **Full term pregnancies**
  - 266 days/38 weeks/9 months
    - 40 weeks if date of last menstrual period is used as the starting date.

- **Due Date**
  - Determined based on date of LMP
  - Only 5% of babies born on their due date.
    - On time/full term: three weeks before due date and two weeks after due date
      - Pre-term: more than three weeks before due date
      - Post-term: more than two weeks after due date
Conception: sperm and egg unite

- Within hours of conception the zygote duplication and division.
  - Single cell is made up of a 23 pairs of chromosomes.
  - Single cell divides into two cells
  - Two cells duplicate and divide into four cells
  - Four cells duplicate and divide into eight cells
  - And continues
One celled zygote eventually multiples into:

- 10 trillion cells by birth
- 100 trillion cells by adulthood

Every single cells carries the exact copy of our genetic instructions.

- This explains why one drop of blood, skin cell or piece of hair can be used to test for DNA.
From one cell stage through eight cell stage:
- The organism consists of stem cells
- Stem cells can develop into any body part.

*Stem Cell Research and Debate
- Stem Cells have potential to develop into many different cell types
- Stem cells can serve as internal repair system
- When stem cells divide, each new cell can remain a stem cell or become a more specialized cell such as muscle cell, red blood cell or brain cell
Stem cells are distinguished from other cell types because

1. they are unspecialized and capable of renewing themselves through cell division
2. under certain physiologic or experimental conditions, they can become induced to become tissue or organ specific cells.

1981: Scientists learned how to derive stem cells from early mouse embryos
1998: Scientists learned how to derive stem cells from human embryos & grow these cells in labs.
Stem Cell Research
GERMINAL PERIOD
DIFFERENTIATION
After the eight cell stage cells begin to specialize.

- Some cells become the placenta and some the fetus.
  - Placenta: organ that surrounds the embryo/fetus and sustains life via the umbilical cord
  - Implantation: process by which the developing placenta connects to the uterus
    - Takes place ten days after conception/takes about a week
Implantation

- 60% of all conceptions fail to implant.
- 70% of all in vitro conceptions fail to implant.
- Women are typically unaware of failed implantation.
  - Period may just arrive a few days later than usual
Successful Implantation

- The organism begins to grow in size because of the placenta is connected to the umbilical cord
  - Bringing in nourishment and carries away waste

- 1st noticeable signs of pregnancy occur after implantation
  - Substances begin to enter a pregnant woman’s body via the placenta
  - Pregnancy tests may now detect the new chemical’s in her urine.
Embryonic Period: 2 - 8 Weeks

- Two weeks after conception, cells begin to show the first sign of body shape and structure.
  - Primitive Streak down the middle of the cell mass that becomes the neural tube.
    - 22 Days after Conception.
    - Neural tube becomes the brain and spinal cord.
  - 4th week: head begins to take shape
    - Eyes, nose and mouth form
    - Blood vessel that will be the heart begins to pulsate
Embryonic Period: 2-8 weeks

- 5th week: arms and legs buds appear
  - Upper arms-Forearms, hands, palms and fingers
  - Legs-feet-toes

- 8th week: fingers and toes separate

*No sexual reproductive system differentiation*
The Fetal Period: 9 weeks-birth

- All body structures are formed
  - Except the sex organs

During fetal period, SRY gene commands male or female sex organs to develop.

By 12\textsuperscript{th} week, genitals are fully formed & begin to send hormones to the developing brain.

*sex hormones cause some differences in fetal brain organization
End of third month: sex organs are present and may be visible in a sonogram/ultrasound.
- 3 month old fetus: Approx. 3 ounces/3 inches

4, 5 and 6 months: cardiovascular system becomes active
- Fetal heartbeat becomes audible by stethoscope

Digestive, Respiratory & Excretory Systems
- continue developing and begin functioning
- Fingernails, toenails, hair and buds for teeth appear
½ million neurons (brain cells) are created per minute during the mid-pregnancy.

Cells begin to move toward designated spots in the brain from the back to the front
- Brain stem-Midbrain-Cortex

- Cortex develops extensively before birth but pre-frontal cortex is not mature until the end of adolescence or later.
Advances in brain development are crucial for survival because the brain regulates basic body functions including breathing.

- **Age of viability:** 22 weeks after conception
  - Fetus can survive outside the mother’s uterus with medical care.
  - 23-26 weeks after conception: 65% of births can be saved by medical care.
Fetal Period: 9 weeks-birth

- During the last 3 months
  - Fetus gains 5 pounds
  - Heart and lungs mature
  - Digestive system begins to function
  - Brain growth makes the cortex develop into layers folding several times to fit in the skull.
Birth

- Approx. 38 weeks-fetal brain signals the release of hormones that trigger the women’s uterus to contract and relax.

- Start of the Labor process:
  - 1st baby: labor lasts approx. 12 hours
  - Subsequent births: labor is shorter
  - Under 30 years: shorter labor
Birth: APGAR SCALE

- Assessment of newborn’s body functioning
- Completed at one minute and five minutes
  - 1. Heart Rate
  - 2. Breathing
  - 3. Reflexes
  - 4. Color
  - 5. Muscle Tone

Score of 0, 1, or 2

5 minute score: 7 or above good
Cesarean Section: C-Section

- Surgical birth where incisions through the abdomen and uterus allow the fetus to be removed quickly.
- 1/3 of U.S. births are by C-Section
- C-Sections are somewhat controversial
  - Less trauma for the newborn
  - Surgery and anesthesia may slow down mother’s recuperation possibly impairing the ability to breast-feed and care for the baby.
Low Birthweight

- Leading cause of infant mortality in the U.S.
- Low Birthweight (LBW)
  - Less than 5.5 lbs
- Very Low Birthweight (VLBW)
  - Less than 3.5 lbs
- Extremely Low Birthweight (ELBW)
  - Less than 2.3 lbs
Causes of Low Birthweight

1. Early birth before 35 weeks of gestation
   - Preterm

2. Slow gain throughout pregnancy
   - Small for gestational age
     - At birth, weight is significantly lower than expected based on time of conception.
       - Nurture is more responsible for low birthweight
         - Maternal illness, exhaustion, infection, malnutrition, drug use
       - Importance of prenatal care weighing the woman at every visit
Prenatal Teratogens

- Teratogens: agents and conditions that impair prenatal development and result in birth defects and even death
  - Can be Structural and behavioral abnormalities
  - Substances: Drugs and pollutants
    - Disease: rubella, measles, chicken pox, aids
    - Pollutants: lead, mercury and pesticides
    - Radiation
  - Conditions: Malnutrition and extreme stress
Some zygotes carry genes that make them more vulnerable.

**Timing of Exposure:**
- Critical period: time when each body part develops
- Sensitive Period: time in which teratogens can interfere with recent growth
- Structural: 1st two months
- Body function and cortex damage: anytime

**Amount of Exposure:**
- Threshold effect: some teratogens are relatively harmless in small doses but become harmful once exposure reaches a certain level.