Beginnings

Heredity, Prenatal Development, and Birth
HEREDITY AND ENVIRONMENT

The “Nature versus Nurture Debate evolves into The interplay between genetics and the environment

Many Factors contribute to the mix

Polygenic
Multifactorial or complex
Examples: Diabetes, heart disease

The Epigenetic Framework
Chromosomes and Genes

- Chromosomes
- Humans 46 (23 pairs usually)
- Genes
- 30,000 in humans
- The Human Genome Project
  - Ethical considerations
  - Emotional relief or distress
  - Lifestyle adjustments
Conception
Reproductive Readiness

- Reproductive cells
  - sperm (active)
  - an ova or gamete
- 23 chromosomes each (leads to meiosis)
- Spermatogenesis
- Oogenesis
The Zygote

- Fertilized egg
- Fertilization in fallopian tube
- Meiosis: Genetic material combines
- Doubling (self multiplication) then differentiation
Determining the Sex of the Child

- 22 chromosomes from each parent are similar in length
- 23rd is either an X or Y
- Half of dad’s sperm contain a Y
- All of ova contain X
- XX female; XY male
- So dad determines sex

Normal Male Karyotype - a female would have two X’s instead of an X and Y
Monozygotic and Dizygotic Twins

- **Monozygotic twins**
  - Single fertilized egg
  - Genetically identical
  - Less common

- **Dizygotic twins**: two eggs and two sperm
  - Share about $\frac{1}{2}$ DNA
  - Most common and increasing (2/3rds)
Genotype and Phenotype

- What you get genetically is not always what you see
- Genotype: genetic potential
- Phenotype: actual physical traits
What Determines the Expression of Genes?

- Additive Pattern: average of genes (height; skin tone)
- Dominant/Recessive Pattern
  - dark hair and dimples (D)
  - red hair and nearsightedness (R)
- Partial dominance or codominance
- Environmental factors (oxygen deprivation, exertion, etc.)
Chromosomal Abnormalities

- Age of mother a major factor
- Down syndrome (20 yr old woman has 1 in 800 chance; 44yr old 1 in 16 chance)
- 1/2 zygotes have abnormal number of chromosomes
- If occurs anywhere but on 21\textsuperscript{st} or 23\textsuperscript{rd} position, organism usually dies
Trisomy 21

- Most common chromosomal abnormality
- An extra chromosome on #21
- Intellectual disability
- Developmental delay
- Distinctive physical features
Sex Linked Chromosomal Abnormalities

- Abnormal number of chromosomes on 23rd pair
- Numerous sex-linked disorders
- XXX, XXY, XO, etc.
- Usually sterility and learning difficulties
- Klinefelter’s Syndrome
Turner’s Syndrome
Genetic Disorders

- Are attributed to specific genes
- Dominant gene linked disorders (Huntington’s Disease)
- Recessive gene linked disorders (Sickle-Cell Anemia, Cystic Fibrosis)
Prenatal Development starts with the Germinal Period

- First 14 days
- Zygote travels to the uterus for implantation
- Cells differentiate
- 60% fail (70% in-vitro)
- Mom unlikely to know she’s pregnant yet
The Embryonic Period

- 3rd through the 8th week
- Major structures begin to form as a result of lots o’ differentiation
- Neural tube that will be the brain and spinal cord is now forming
- Proximodistal & cephalocaudal patterns emerge
- 20 percent fail
- Human appearance

Photo Courtesy Lunar Caustic
The Fetal Period

- 9th week until birth
- Early on we are developing genitalia, hair & nails, digestive system etc
- End of 3rd month, all the parts are present
- Later on we are working on reflexes like sucking and swallowing
- Age of viability (24 weeks)
- 7-9th month gain 5 lb, 7 inches
- 5 percent fail
Teratology and Risks in Prenatal Development

- Timing of exposure (1st 3 months critical)
- Amount of exposure (more is worse)
- Genetics (mom’s and junior’s)
- Gender
- Environmental pollutants like lead and mercury are very hazardous
- Serious issues if mom contracts Rubella or has HIV
- Drugs like heroin, cocaine, marijuana
Critical Periods of Development
Alcohol

- Most common teratogen
- Vulnerability during the 2\textsuperscript{nd} month of development
- Moderate to heavy drinking or a single binge (varies)
- Fetal Alcohol spectrum disorder
Fetal Alcohol Spectrum Disorders

- Learning difficulties
- Impaired motor skills
- Flattened nose
- Widely spaced eyes
- Small heads

- Long term psychosocial problems
- ARND and ARBD
  - Neurological damage
  - Kidney, bone, heart problems
Tobacco Use

- Our 2\textsuperscript{nd} most popular teratogen
- Fetal growth restriction
- Low birth weight
- Placenta previa
- SIDS
- Neurological problems
  - (ADHD)
Minor Complications of Pregnancy

- Nausea, heartburn, gas
- Hemorrhoids, backache, leg cramps
- Insomnia, constipation
- Shortness of breath
- Varicose veins
- Cure? Delivery!!!!
Major Complications

- Severe vomiting
- Threatened abortion
- Toxemia
- Preeclampsia; may lead to eclampsia
- Ectopic (tubal) pregnancy
- Maternal mortality
  - Infection
  - Excessive bleeding
Problems of Newborn

- Low Birth Weight <5.8 pounds (8% U.S.)
  - Tobacco, alcohol, use; poor nutrition;
- Preterm early < 37 weeks (13%) often a contributor to low birth weight
- Fetal Anoxia – oxygen deprivation during delivery
Child birth methods: Start with Dick-Read Method

- Dick-Read: "Childbirth Without Fear: The Principles and Practice of Natural Childbirth"
- Pain comes from fear
- Education reduces fear

- Pregnancy Today - Childbirth Without Fear: The Teachings of Grantly Dick-Read
The Lamaze Method

- From Russia to the US in the 1950s
- French obstetrician Fernand Lamaze
- Teach control
- Breathing, relaxation and focal point
- Have a coach
- Manage rather than eliminate pain
Other Methods

- LeBoyer: gentle birthing
- Soft lighting and a soothing environment
- Out of hospital births (1 percent of births) usually assisted by a midwife
  - $\frac{2}{3}$ are Home births
  - $\frac{1}{3}$ occur in birthing centers
  - Moms are usually Caucasian and experienced
Stages of Delivery

- **First Stage**
  - Longest Stage
  - Uterus contracts & Cervix dilates
  - Discharge of blood or amniotic fluid

- **Second Stage**
  - Baby through birth canal
  - 10-40 minutes
  - Head normally delivered first
  - Mother pushes and relaxes
  - Episiotomy
Stages of Delivery

- Third Stage
  - Placenta or afterbirth is delivered
  - Episiotomy sutured
  - Relatively painless
Assessing the Newborn

- The APGAR
  - Heart rate, respiration, muscle tone, reflex response, and color
  - Score 0-2 on each
  - Total score <5 is a cause for concern

Doctors also frequently check the Babinski reflex
The Neonatal Behavioral Assessment Scale (NBAS)

Tests neurological condition/behavior in four areas:

- Motor development
- Interactive behavior
- Response to stress
- Physiological control