Chapter 3: Forming a New Life: Conception, Heredity, & Environment

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Forming a New Life: Conception, Heredity, and Environment

- Becoming Parents: How Conception Occurs
- Infertility
- Mechanisms of Heredity
- Genetic and Chromosomal Abnormalities
- Nature and Nurture: Influences of Heredity and Environment
- Some Characteristics Influenced by Heredity and Environment
How Conception Occurs

• Changing theories of conception
  – Hippocrates, father of medicine, believed fetus results from male and female seeds
  – Aristotle, philosopher, believed woman was only receptacle of child formed by the sperm
    • Male babies were natural; females result only if development was disturbed
    • Sperm were miniature versions of people
  – Ovists, inspired by Dr. Harvey, said ovaries contained tiny, already formed humans activated by male’s sperm
  – German anatomist Wolff demonstrated embryos not preformed and both parents contribute equally to new being
• **How fertilization occurs**
  - Sperm and ovum: gametes or sex cells
  - Sperm and ovum unite: zygote (which then duplicates)
  - At birth, female has 2 million immature ova in two ovaries
  - Ovulation occurs approximately every 28 days
  - Ovum travels from ovary, down fallopian tube, to uterus
  - Sperm: several hundred million produced daily, released during ejaculation
    • Travel up through the vagina, through the cervix, into the fallopian tubes
  - Fertilization generally occurs in fallopian tube, travels down to uterus, embeds into uterine wall
  - Fertile window from 6th – 21st day
  - Sperm absorbed by woman’s white blood cells
  - Ovum passes through the uterus and exits through the vagina
Sperm cell

- Acrosome
- Nucleus
- Centrioles
- Mitochondria
- Head
- Midpiece
Mitosis: zygote, after conception, 1 cell division
Fetal Development
Infertility

- Infertility
  - 7% of US couples infertile
    - Inability to conceive baby after 12 months of trying
  - Women’s fertility declines in late 20’s, substantial decreased during 30’s
  - Men’s fertility less affected by age but declines significantly by late 30’s
• **Causes of infertility**
  
  – *Most common reason in men is production of too few sperm*
    
    • Sperm count of lower than 60 – 200 million per ejaculation makes conception unlikely
    
    • Also, sperm unable to swim well enough to reach cervix
  
  – *Most common reason in women is blockage of fallopian tubes*
    
    • ½ cases are blocked by scar tissue from sexually transmitted diseases (STIs)
    
    • Non-production of ova
    
    • Mucus in cervix
    
    • Disease of uterine lining
    
    • Over 30, deterioration in quality of ova
• **Treatment for infertility**
  – Hormone treatment
  – Drug therapy
    • Results in multiple births
  – Surgery
  – Men undergoing fertility treatments are at increased risk of producing sperm with chromosomal abnormalities
    • Daily coenzyme Q10 increases sperm motility
  – Pregnancies occurring after a year or more of trying have more pre-term births, low birth-weight, cesarean deliveries
• **Alternative ways to parenthood**

  – **2003 1% babies in US born were assisted with technology**
    
    • **In vitro fertilization**
      – Most common
      – Fertility drugs given to increase production of ova
      – Harvest mature ovum (surgical removal)
      – Fertilized in laboratory dish
      – Implanted in woman’s uterus

    • **Artificial insemination**
      – Injection of sperm into vagina, cervix, or uterus
      – For low sperm count

    • **GIFT:** Gamete intrafallopian transfer and zygote intrafallopian transfer
      – 35% live births as result of assistive technology

  – **Surrogate motherhood**
    
    • Several options, laws protecting fetus
Mechanisms of Heredity

- **Genetic Code**
  - Basis of heredity is DNA
  - Pairs of bases
    - Adenine (A)
    - Thymine (T)
    - Cytosine (C)
    - Guanine (G)
  - Chromosomes are coils of DNA
    - **Each cell has 23 pair = 46 chromosomes (if normal)**
  - Genes reside on the chromosomes
    - Functional units of heredity
Human Chromosomes
Karyotype
Deoxyribonucleic acid
What Determines Sex?

• All ovum are X
• Sperm can carry X or Y
• If ova fertilized by X sperm: X + X = girl*
• If ova fertilized by Y sperm: X + Y = boy*
Patterns of Genetic Transmission

• Everyone receives a pair of alleles for a characteristic, one from each parent
  – When alleles are the same, homozygous
  – When alleles are different, heterozygous

• Dominant inheritance*
  – Heterozygous, only one of the allele pair will be expressed

• Recessive inheritance*
  – Recessive allele from both parents
Patterns of Genetic Transmission (cont)

• Polygenic inheritance
  – Multiple genes on different chromosomes create trait

• Mutations

• Multifactorial transmission: combination of genes and environment involved in expression of a trait

• Genotypes *
  – Underlying genetic makeup

• Phenotypes *
  – Observable characteristic
Genetic and Chromosomal Abnormalities

• Dominant or recessive inheritance of defects
• Defects transmitted through recessive inheritance more likely to be lethal at early age
• Defects transmitted through dominant that are harmful occur later in life
  – If occurred in early life, wouldn’t reach age of reproduction
• Sex-linked inheritance of defects
  – Almost always appear in males
  – Females, normal dominant gene on X chromosome from father overrides defective gene on mother’s X chromosome
• Chromosomal abnormalities
  – Errors in cell division, meiosis
    • Klinefelter syndrome XXY
    • Turner syndrome XO
Frontal baldness absent
Tendency to grow fewer chest hairs
Breast development
Female-type pubic hair pattern
Poor beard growth
Narrow shoulders
Wide hips
Small testicular size
Long legs

Short stature
Low hairline
Shield-shaped thorax
Widely spaced nipples
Shortened metacarpal IV
Small finger nails
Brown spots (nevi)

Characteristic facial features
Fold of skin
Constriction of aorta
Poor breast development
Elbow deformity
Rudimentary ovaries
Gonadal streak (underdeveloped gonadal structures)
No menstruation
low posterior hairline
webbed neck and extra skin
wide-set nipples
discolored spots on skin
swollen hands

swollen feet

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• **Down syndrome**
  
  – Autosome cell division
    • Trisomy 21 (three chromosomes 21)
    • Translocation (inherited)
  
  – 1 in 700 births
  
  – Risk is greatest for older parents: mother over 35
  
  – Extra chromosome comes from mother 95% of the time
flattened nose and face, upward slanting eyes,

single palmer crease, short fifth finger that curves inward

widely separated first and second toes and increased skin creases

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Genetic Counseling and Testing

• Helps prospective parents assess their risk of genetic or chromosomal defects
  – Parents who have a child with a genetic defect
  – People with family history of hereditary illness
  – Ethnic groups at higher-than-average risk of certain diseases
    • Tay Sachs

• Blood work, urinalysis, skin sample

• Implications
Nature and Nurture: Influences of Heredity and Environment

- **Heritability**
  - Statistical estimate of contribution of heredity to individual differences in a specific trait, eg, the color of my skin will resemble my family’s color with variation, depending on prior genes: heritability
  - Does not tell us how the traits develop
  - Does not tell us influence of environment
  - Adoption studies show influence of environment
    - Obesity
  - Twin studies
    - Monozygotic (one zygote): twice as genetically similar than dizygotic twins
    - Dizygotic (two zygotes): no more similar than siblings born years apart
Nature and Nurture: Influences of Heredity and Environment (cont)

- Heredity and environment work together
  - Genes and experiences are intertwined, not working independently as once believed
    - Cognition and retardation
    - Role of SES
    - Role of neighborhood
    - Role of educational opportunities
Nature and Nurture: Influences of Heredity and Environment (cont)

• **Genotype-environment correlation**
  
  – Passive correlations: parents who provide the gene also provide the environment, such as music, child grows up with music
  
  – Reactive correlation: child with certain genes evokes different responses from adults, such as music, parents not talented but respond to child who is
  
  – Active correlation: child gets older and chooses own activities, chooses experiences consistent with genes, such as musical talent, takes music classes, goes to concerts
Nature and Nurture: Influences of Heredity and Environment (cont)

- **What makes siblings so different?**
  - Nonshared environment accounts for most of differences*
    - Genetic differences lead children to need different kinds of stimulation
    - Genetic differences lead children to respond differently to a similar home environment
      - One child may have larger reaction to arguments and aggression within a family
    - Parents treat children differently as a result of temperament, economics, stresses present
    - Illnesses, accidents
    - Events outside the home: teachers, peers
  - Shared environment
    - Home, people in the home, activities family engages in
  - How child perceives an event influences their response
  - Children mold their environments by the choices they make
    - Influenced by their genes!
Nature and Nurture: Influences of Heredity and Environment (cont)

• Epigenesis: environmental influence on gene expression
  – Diabetes
  – Cancer
  – Heart disease
Characteristics Influenced by Heredity and Environment

1. Physical and physiological traits
   - Medical disorders more concordant in monozygotic twins
     - High blood pressure, heart disease, stroke, rheumatoid arthritis, peptic ulcers, epilepsy
   - Life span
   - Obesity, multifactorial condition: BMI 95th % for age and sex
     - 40 – 70% of risk is genetic
     - Chromosome 10, gene GAD2, controls appetite but abnormal gene stimulates hunger and overeating
     - Kind and amount of food is environmental
     - Physical activity and exercise is environmental
Characteristics Influenced by Heredity and Environment (cont)

2. **Intelligence (polygenic)**
   - Strong heredity influence on general intelligence
   - Less heredity influence on:
     - Memory
     - Verbal ability
     - Spatial ability
   - Adopted children’s IQ closer to biological parents’ than adoptive parents’
   - Monozygotic twins closer than dizygotic twins
   - Genetic influence increases with age
     - Shared family environment is strong influence on young children but a diminishing influence on adolescents and adults
   - Nonshared environment is influential throughout life and is primarily responsible for changes in cognitive performance
3. **Personality**

- Genes directly linked with specific personality traits:
  - Neuroticism, which may contribute to depression and anxiety
  - 40 – 50% heritability
  - Shared environment is not an influence on personality

- **Temperament (Thomas and Chess)**
  - Inborn
  - Consistent over time but responsive to parental responses and experiences
  - Genetic influences:
    - Activity level
    - Sociability
    - Emotionality

- Religiousness is both genetic and environmental
  - Parenting and family life has strongest influence in childhood
  - Genetic influences more predominant from adolescence on
4. Psychopathology

- Strong hereditary influence:
  
  - Schizophrenia (neurological disorder)
    - Loss of contact with reality, hallucinations, delusions
    - Multifactorial causes, no single gene
      - May be lack of chemical reelin
      - May be prenatal insults
      - Urban areas more prevalent
      - Late winter, early spring births increase risk
      - Obstetric complications
      - Poverty, severely deprived as a result of war or famine
      - Fetal malnutrition
      - Advanced paternal age (father over 30 years)
    - 10 times greater among siblings and children of schizophrenics than general population
    - 80 – 85% heritable
  
  - Autism
  
  - Depression

- Inherited tendency can be triggered by environmental factors
“Parenting is a two-way street. As you take them by the hand, they will take you by the heart.”

-Judy Ford