CHAPTER 4: HUMAN DEVELOPMENT: BIRTH TO SIX

Motor development:

• **Basis:**
  • Movement
  • Posture
  • Balance
• Motor development is integral to acquiring:
  • Concrete knowledge
  • Producing speech
  • Exploring the environment
  • Carrying out daily self-help activities
  • Socializing with others
MOTOR DEVELOPMENT: (CONT)

- Normal physical development allows:
  - Organized movement
  - Purposeful movement
  - Efficient movement
- How does physical development occur?
  - Neurological maturation (historic view)
  - Systems model based on biomechanical theory
    - More ecological approach
    - Includes influence of the environment
  - The task at hand
  - Psychological processes
    - Motivation
    - Personality
  - Interaction between musculoskeletal and neurological maturation
GROSS MOTOR DEVELOPMENT

- **Muscle control progresses:**
  - Cephalo-caudal
  - From gross motor to fine motor
  - Proximal to distal
  - Reflex to intentionality
  - Simple to complex
- Head control
  - Supine to prone
- Shoulder control
  - Allows pivoting and turning over by 4th or 5th month
- Trunk control
  - Sit up independently at 6 months
  - Get into and out of sit at 8 months
- Hip control
- Lower body control
- Refinement
WINDOWS OF ACHIEVEMENT

Sit without support: 4 – 9 months
Stand with assistance: 5 – 11 months
Hand and knee crawling: 5 – 13 months
Walking with assistance: 6 – 14 months
Standing alone: 7 – 17 months

Walking alone: 9 – 17 months
MOTOR DEVELOPMENT: FINE MOTOR SKILLS

- Eye contact
- Facial expression
- Reaching
- Grasping
  - Palmar
  - Pincer
- Handedness
  - Reflection of hemisphere dominance
LANGUAGES DEVELOPMENT

- Communication
- Speech
  - sounds
- Language
  - Communicating idea, wish, desire, need, emotion
- Receptive language: usually higher ability
  - What is understood
- Expressive language: usually lower than receptive language
  - What is said
- Innate ability for language
- Acquired in universal pattern
- Modeled by more competent speakers, repeated practice of sounds and words
LANGUAGE FORM

• Three aspects of language
  • Phonology: study of speech sounds
  • Syntax: rules of language, grammar
  • Morphological development: word structure and word parts, such as prefixes and suffixes
LANGUAGE FORM

- Phonology
- Syntax
- Morphological development
PHONOLOGICAL DEVELOPMENT

- Cooing - vowel sounds
- Babbling - C/V (consonant/vowel)
- Sound production
  - D, t, k, m, h (8 months)
  - G, n, b
  - W, s
  - P, s, k, z (two years)
  - F, ts, j, l, r, pw, bw
  - Fw, kw, pl, nts, nd, ps, ts
  - Dz, sp, st, sn, sl
  - Tw, sk, sm, bl, kl, gl, br, tr, dr, kr, gr (4 years)
  - V, fr, sr, pr, fl (5 years)
- Ma-ma, da-da, pa-pa nonspecifically
- Mama dada papa specifically
- Single words, objects
- Two-word sentences after approximately 30 – 50 words
**SYNTAX**

- Simple two-word sentences include noun-verb, verb-noun, and noun-noun
- “Wh” questions are challenging
  - Who, what, where, when, how, why

**MORPHOLOGICAL DEVELOPMENT**

- Morpheme: smallest part of a word that has meaning
- Mean length of utterance (MLU)
  - More than three, have morphological inflections in phrases
EXPRESSIVE LANGUAGE

- **Semantics**
  - What words mean

- **Overextension**
  - All blue cars are Mommy’s car

- **Underextension**
  - Categories are too narrow: only my red blanket is my blankie; all other blankies are something else
FACTORS AFFECTING LANGUAGE DEVELOPMENT

- Developmental disabilities
- Cleft lip/cleft palate
- Oral-structural anomalies associated with Down syndrome
  - Influence phonological production
- Cerebral palsy
  - Reduced respiratory capacity
- Autism
  - All areas of language affected
- Language deficits often inherited (50%)
- Culture and sociocultural factors influence rate of language acquisition
- Parenting influences language acquisition
  - Motherese is correlated to language development
- Children with disabilities may require structured opportunities to facilitate language development
  - With children whose language is developing typically
COGNITIVE DEVELOPMENT

• How we acquire knowledge
• Early experiences are critical to acquiring knowledge and sculpt:
  • Perception
  • Selective attention
  • Learning
  • Memory
  • Language
  • Personality
  • Cognition
• Brain is plastic and can create alternative routes to adjust to auditory and visual experiences
• Repeated exposure to stimuli molds a response and pathways that have not been able to respond, drop out and die
THEORIES OF COGNITION

• **Piaget: Developmental approach**
  • Four distinct stages:
    • Sensorimotor
      • Six substages
        • Reflex activity
        • Primary circular reactions
        • Secondary circular reactions
        • Coordination of secondary schemes
        • Tertiary circular reactions
        • Mental combinations
  • Object permanence
  • Attachment
    • Strong indicator of memory in infants

• Preoperational
  • Representational thought
  • Symbolic ability
  • Egocentrism
  • Irreversibility

• Concrete operations
  • Decentration
  • Reversibility
  • Logic
  • Conservation
  • Relational thinking (shorter, longer)
  • Hierarchical relationships

• Formal operations
  • Flexible thinking
  • Abstract thinking
SOCIAL EMOTIONAL DEVELOPMENT

- Emergence of individual emotions and personality is result of and influence on dynamic relationships with others and the environment
- Adapting to community norms that govern living within a society
- Social development:
  - Observable behavior
- Emotional development:
  - Takes place under the skin
  - Influenced by language and cognitive development
- Influences on social-emotional development
  - Heredity
  - Culture
  - Economics
  - Community
SOCIAL EMOTIONAL DEVELOPMENT (CONT)

- Maternal stress and anxiety increases cortisol that crosses the placenta that affect hormonal and brain development
- When infants experience prolonged subtle forms of emotional deprivation (when mothers are depressed) they experience a dampening of their own emotions
  - Fail to gain weight
  - Lethargic
  - Development is compromised
- Environmental factors that place parents at risk of being inadequate nurturers of secure attachment:
  - Substance abuse
  - Child abuse
  - Underage pregnancy
  - Low socioeconomic status
  - Economic stressors
  - Poverty
  - Infant prematurity
  - Overcrowding
  - Absent fathers
SOCIAL EMOTIONAL DEVELOPMENT (CONT)

- Parenting affects development:
  - Emotional resiliency develops when mothers are primarily positive
    - Young children learn to regulate their emotions and their impulses
  - Coercive parenting patterns lead to harsh and inconsistent consequences, leading to later social problems and emotional depression
VYGOTSKY: CRISIS OF THREE: DEFIANT BEHAVIOR

- Negativism
- Stubbornness
- Obstinancy
- Willfulness
- Protest
- Devaluation
  - Calls stupid or dumb
- Despotism
  - Wants complete power over those around
CHILDREN WITH DISABILITIES

• Impairment in one area can significantly alter the ability of child to initiate or respond to interactions that build or maintain social relationships
SOCIAL PLAY

- Play is the medium that infants and toddlers acquire and execute social relationships
- Play is the work of young children
- Economic stability in a family contributes to more social contacts
TYPES OF PLAY (COGNITIVE)

- Solitary play
  - Isolation and independence
- Spectator play (onlooker play)
- Parallel play
- Associative play
  - Lacks organization
  - Toys shared without regard for group’s wishes
- Cooperative Play
- Games with rules
  - 6 years of age
- Children learn:
  - Truth
  - Honesty
  - Fair play
  - Self-control
  - Leadership skills
SELF-HELP DEVELOPMENT

- AKA Adaptive skills
  - Independent feeding
  - Dressing
  - Toileting
  - Personal responsibilities
INDEPENDENT EATING AND DRINKING SKILLS

• Sucking reflex
• Solid pureed foods
• Table foods
• Dramatic change around 8 months
• Drink from cup
• Feed themselves finger foods
• Age 2 independent eaters and drinkers
  • Use of spoon
  • More narrow range of food preferences
DRESSING SKILLS

- Pull socks off
- Assist in dressing
- Pull pants up and down
- Zippers
- Dress self by 3 years
TOILETING SKILLS

• Muscle control not fully developed until 2 years of age
• Bowel movements may cause fear in child
• Not unusual to still use diapers at three years of age, especially if any other delays exist
PERSONAL RESPONSIBILITY

- Pick up their toys
- Routines
- Wipe up spills
- Clear the table
- Dirty clothes in a hamper
Children With Disabilities

Chapter 4: Birth Defects and Prenatal Diagnosis
BIRTH DEFECTS AND PRENATAL DIAGNOSIS

Upon completion of this chapter, the student will:

1. Understand the uses and limitations of noninvasive prenatal maternal blood screening for birth defects

2. Be knowledgeable regarding the indications for, and limitations of, first- and second-trimester evaluation of birth defects using the techniques of ultrasound, fetal MRI and ECG

3. Be aware of techniques of amniocentesis and chorionic villus sampling to determine when these invasive diagnostic tests may be indicated

4. Be familiar with alternative reproductive techniques (IVF) and understand when couples might benefit from such technologies

5. Learn about new noninvasive prenatal diagnosis technologies being explored

6. Understand the psychosocial needs of families who are at risk
BIRTH DEFECTS AND PRENATAL DIAGNOSIS

- 3% of births result in a child with a birth defect or genetic disorder
- Circumstances can increase risk
- Most affected newborns are born to couples unaware they are at risk and have no family history
BIRTH DEFECTS AND PRENATAL DIAGNOSIS

• Noninvasive prenatal maternal blood screening
• First-semester evaluation of birth defects
  • Ultrasonogram
  • Fetal magnetic resonance imaging
  • Echocardiography
• Second-trimester evaluation of birth defects
• Invasive diagnostic tests
  • Amniocentesis
  • Chorionic villus sampling
• Alternative reproductive techniques
  • In vitro fertilization
• Psychosocial needs of families at risk for having children with genetic disorders or birth defects
PRENATAL DIAGNOSIS AND SCREENING

• Gives parents opportunity to gain information about fetus
• Gives parents opportunity to examine a range of family planning options
• Screening can occur before pregnancy or during pregnancy
GENETIC ASSESSMENT

- 20,000 genetic disorders have been identified
  - Genetic testing available for 2,000 genetic disorders
- Ethnic background
  - Specific ethnic backgrounds have higher chance of certain gene mutations associated with genetic disorders
- Review of medical and pregnancy history
- Extended family history
- Presence of family birth defects and genetic disorders
- Unexplained infant deaths
- Recurrent pregnancy losses
- Maternal medication use
- Occupational or teratogen exposure
- Carrier screening
  - Autosomal recessive
  - High morbidity (disease) and mortality (death)
GENETIC ASSESSMENT

• Genetics Home Reference
• National Library of Medicine supported database
• National Organization for Rare Disorders (NORD)
  • www.rarediseases.org
• Genetic Alliance
  • A clearinghouse for information and support groups for genetic disorders
  • www.geneticalliance.org
SCREENING EVALUATIONS DURING PREGNANCY

- **First Trimester**
  - Ultrasound
    - Establishes viability
    - Determines number of fetuses
    - Confirm placenta position
    - 11 – 14 weeks can measure nuchal translucency (transparency of the fluid-filled cavity at the nape of the fetus’s neck)
      - Increased nuchal translucency is associated with adverse outcomes
        - Congenital heart disease
        - Fetal anomalies
        - Fetal death
    - Down syndrome: abnormal Doppler flow in ductus venosus and tricuspid regurgitation (signs of congenital heart disease)
  - Maternal serum (blood) screening
  - Screening for disorders common in specific ethnic groups
    - Cystic fibrosis
    - Sickle cell anemia
    - Tay Sachs
  - Chorionic villus sampling
SCREENING EVALUATIONS DURING PREGNANCY

• First-Trimester
  • Maternal serum screening
    • Free beta hCG and PAPP-A at 10 – 14 weeks combined with ultrasound identifies 87% fetuses with Down syndrome
      • <5% false-positive
    • Extreme variations of free beta hCG and PAPP-A can indicate:
      • Low birth weight
      • Stillbirth
      • Fetal loss
      • Early delivery
  • Cell-free DNA in plasma of pregnant woman
    • Fetal DNA can be detected in background of maternal DNA, assessment fetal chromosomal abnormalities
    • Further work needs to be done before technique is widely available for clinical use
SCREENING EVALUATIONS DURING PREGNANCY

• **First trimester**
  • Chorionic villus sampling
    • Performed 10 – 12 weeks
    • Use ultrasound to guide suction through small catheter passed through cervix or aspiration via needle inserted through abdominal wall and uterus
    • Minute biopsy of chorion (outermost membrane surrounding embryo)
      • Consists of rapidly dividing cells
    • Detects chromosomal abnormalities
    • Use for enzyme assay for inborn errors of metabolism
    • Use for molecular DNA analysis to identify specific mutations that cause genetic diseases
    • Does not detect neural tube defects such as spina bifida
    • Safest invasive prenatal diagnostic procedure
      • 1% risk of procedure-related pregnancy loss
      • If performed after 10 weeks, no increased risk of causing fetal anomaly
CHORIONIC ASPIRATION

Transcervical Chorionic Villus Sampling

Catheter taking tissue samples

Ultrasound Transducer

Uterus

Placenta

Fetus
TRANSABDOMINAL CVS
SCREENING EVALUATIONS DURING PREGNANCY

Second Trimester

- Maternal serum screening
  - Screening sensitivity is improving but is not diagnostic
  - AFP (low), hCG, uE3, and Inhibin A has improved detection of Down syndrome, 80% with 5% false-positive rate
  - When combined with other indicators including maternal age, weight, race, diabetic status and number of fetuses, can assess risk for:
    - Neural tube defects (spina bifida and anencephaly)
    - Abdominal wall defects (gastroschisis and omphalocele)
    - Trisomy 18
  - AFP high levels can be associated with multiple fetuses, gestational age greater than anticipated or higher risk for preterm delivery, stillbirth, or intrauterine loss
  - If serum screen suggests increased risk for Down syndrome, trisomy 18 or trisomy 13, diagnostic testing is recommended
SCREENING EVALUATIONS DURING PREGNANCY

Second-trimester

- **Ultrasound**
  - 18 – 20 weeks detects 60% of major structural anomalies
  - Can be used to diagnose neural tube defects and abdominal wall defects (previously screened for by second-trimester serum testing)
  - High resolution ultrasound revolutionized identification of fetal anatomic abnormalities
  - Can be used to diagnose:
    - Facial clefts
    - Renal anomalies
    - Skeletal anomalies
    - Hydrocephalus
    - Heart defects
    - Other malformations
  - Does not replace amniocentesis and CVS
ULTRASOUND
SCREENING EVALUATIONS DURING PREGNANCY

Second trimester

• **Amniocentesis**
  - Performed at 15 – 18 weeks
  - Ultrasound used to guide procedure
  - Needle inserted below mother's umbilicus, through abdominal and uterine walls
  - 1 – 2 ounces of amniotic fluid aspirated
  - Fetal urine replaces fluid in 24 hours
  - Pregnancy loss when performed before 14 weeks increases, higher rates of musculoskeletal deformities (club foot), and greater risk of fluid leakage
  - CVS preferred first trimester
SCREENING EVALUATIONS DURING PREGNANCY

MAGNETIC RESONANCE IMAGING

- Used approximately 17 weeks
- No sedation
- No known risks at this time
- MRI of central nervous system can demonstrate presence:
  - Corpus callosum
  - Chiari malformation of the brain
  - Cause of enlarged ventricles (hydrocephalus)
  - Ultrasound identifies the risk, MRI confirms

http://www.youtube.com/watch?v=TiG2nbsc-gc
http://www.youtube.com/watch?v=SFY5COYwIXU
https://www.youtube.com/watch?v=ijhLPWBzziA
https://www.youtube.com/watch?v=w7rh_mdL71Q
https://www.youtube.com/watch?v=aHEi_31IYhg
https://www.youtube.com/watch?v=RcKfgBqU0H0
SCREENING EVALUATIONS DURING PREGNANCY

Second trimester

- **Fetal Echocardiography**
  - Congenital heart disease (CHD) most common anatomical abnormality, contributing to 1/3 of congenital anomaly deaths in childhood
  - Targeted ultrasound performed 18 – 22 weeks
  - Three- and four-dimensional studies
  - 10 – 15% of infants with CHD have underlying chromosomal abnormality
DIAGNOSTIC TESTING OF FETAL CELLS

• CVS
• Amniocentesis
  • To test for chromosomal analysis
• Biochemical analysis
  • For inborn errors of metabolism or DNA analysis
    • For fragile X syndrome or cystic fibrosis
• Fluorescent in situ hybridization (FISH)
  • Short pieces of DNA (called DNA probe) of known sequence can attach to a unique region on a chromosome
  • FISH used to identify specific chromosomes or indicate small deletions of a defined region of a specific chromosome
  • FISH used to detect microdeletions
  • Discover of certain CHDs should prompt consideration of FISH analysis to detect 22q11.2 deletion associated with VCFS/DiGeorge Syndrome, 1 in 4,000 live births
“The most sophisticated prenatal diagnostic technology cannot guarantee the birth of a typical child. Most of the disorders that cause developmental disabilities in the absence of structural malformations are not currently amendable to prenatal diagnosis.”
PREVENTION AND ALTERNATIVE REPRODUCTIVE CHOICES

- Receive early prenatal care
- Avoid alcoholic beverages and tobacco
- Minimizing unnecessary medication
- Avoid exposure to infection
- Avoid excess vitamin A
- Avoid frequent consumption of fish known to have elevated mercury content
- Take 0.4 mg of folic acid of childbearing age 3 months before attempted conception
- Maternal diabetes and lupus increases risk to fetus
- Certain medications (e.g. anti-epileptic drugs) increase risk of birth defects
  - In utero repair of myelomeningocele reduces hydrocephalus and increased likelihood of future independent ambulation
  - Maternal morbidity (disease) is risk
- Maternal PKU is at risk of child with microcephaly and intellectual disability if she does not maintain diet during pregnancy

*Always weigh risk of procedures with outcome to fetus and mother.*
ASSISTED REPRODUCTION TECHNOLOGY (ART)

- Mendelian genetic disorders inherited as autosomal recessive (two carrier parents)
- X-linked recessive (with a carrier mother)
- Autosomal dominant (one parent affected)
- Parent with balanced chromosome translocation at risk for unbalanced genetic material
- Artificial insemination using donor sperm
- In vitro fertilization with donor egg
- In vitro fertilization with donor egg and donor sperm
- Use of proteomics (methodology for measuring proteins) may assist in identifying embryos with highest implantation potential
- Evaluation of secretome, proteins embryo produces and secretes into environment may provide clues
- Intracytoplasmic sperm injection (ICSI) for infertile males with low sperm count or poor sperm motility
  - Sperm harvested, cytoplasmic portions of sperm removed, nucleus of sperm introduced into harvested egg by microinjection and blastocyst transferred into uterus
Assisted Reproduction Technology (cont)

- Genetic causes of male infertility assisted by ICSI:
  - Microdeletion within fertility-associated regions of the Y chromosome
  - Certain cystic fibrosis mutations
  - Klinefelter syndrome
  - 1% of conceptions as a result of ICSI result in aneuploidy (extra X or Y chromosome)
  - 6.5% malformation rate secondary to the procedure
PREIMPLANTATION GENETIC DIAGNOSIS (PGD)

• For couples who are high risk of child with genetic disorder
ASSISTIVE REPRODUCTIVE TECHNOLOGIES: THE COSTS

- Costly physically
- Costly emotionally
- Costly financially
- Health insurances rarely cover
- Risk of multiple gestations
- Fetal reduction
- A suggested association between ART and imprinting (epigenetic) disorders such as Beckwith Weidemann Syndrome
PSYCHOSOCIAL IMPLICATIONS

• Choices can be overwhelming
• Health care professionals often avoid difficult preliminary discussions
• Some parents find advance knowledge helpful in preparing before birth
• Some parents terminate pregnancy
• Some parents grieve entire pregnancy
• Health care professions must focus on the family’s psychosocial needs as well as the clinical information the parents are requesting
• A mother giving birth to a child with special needs or to a child who does not survive can be devastated
• ASSUMPTIONS AS TO WHAT A FAMILY SHOULD OR SHOULD NOT DO MUST BE AVOIDED
• Support groups and/or counseling can be helpful
Children with Disabilities

Chapter 5: Newborn Screening: Opportunities for Prevention of Developmental Disabilities
NEWBORN SCREENING

Upon completion of this chapter, the student will:

1. Understand the rationale for newborn screening
2. Understand the difference between a screening test and diagnostic test
3. Be familiar with the types of screening tests available
4. Understand the limitations and pitfalls of screening
NEWBORN SCREENING

- Screening tests screen for but do not diagnose conditions
- All screening tests produce false-positive results
- Some screening tests produce false-negative results
- A diagnostic test definitively confirms or excludes the presence of a disease or condition
WHY SCREEN NEWBORNS?

• Numbers of conditions tested for varies state to state

• American College of Medical Genetics recommends universal screening for 29 specific core conditions and 25 specific secondary conditions
  • Conditions are serious
  • Conditions are identifiable
  • Conditions are treatable
MEDICAL CONDITIONS THAT CAN BE IDENTIFIED BY NEWBORN SCREENING

• **Endocrine disorders**
  • Congenital hypothyroidism: produces inadequate thyroid hormone
    • 1 in 3,000
    • Early treatment replacing the hormone prevents severe growth problems and abnormal brain development, resulting in serious lifelong cognitive disability

• **Infectious diseases**
  • Three states screen for HIV
    • All pregnant women are offered HIV screening to begin treatment and reduce risk of mother to fetus transmission
  • Two states screen for toxoplasmosis gondii (1 in 1,000 to 1 in 8,000)
    • Untreated results in multisystem disease
      • Neurologic complications such as seizures
      • Visual impairments
      • Intellectual disability
MEDICAL CONDITIONS THAT CAN BE IDENTIFIED BY NEWBORN SCREENING (CONT)

• **Immune disorders**
  • SCID (Severe Combined Immunodeficiency: group of disorders leading to early childhood death (bubble boy))
    • 3 states screen
    • Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children recommend adding this to the 29 others currently screened for
    • Children benefit from stem cell transplants

• **Metabolic disorders**
  • Inborn errors of metabolism
  • Often accumulation of abnormal substances, or metabolites, in body fluids and tissues
    • Contributes to abnormal functioning of proteins (enzymes)
    • PKU: 1 in 10,000
      • Amino acid disorder
      • Genetic mutation
      • Can’t process certain proteins (meat; dairy)
      • Protein-restricted diet
      • Not treating causes severe damage to CNS
    • MCAD: 1 in 15,000
      • Organic acidemia, alactosemia, biotinidase deficiency
NEWBORN SCREENING

- **How is newborn screening done?**
  - Blood, heel prick
  - Tandem mass spectrometer
    - Improves detection rate and lowers false positives
  - Second tier testing when positive

- **What should be done when a child has a positive newborn screen?**
  - Notify infant’s doctor, treatment initiated when available, and genetic counseling offered

- **What happens to children with confirmed disease?**
  - Medications, supplements, dietary changes
  - Genetic counseling, family planning

- **What is the risk of developmental disability in children with confirmed disease?**
  - Early detection resulted in fewer hospitalizations, shorter hospital stays, 60% fewer medical problems, scored significantly higher on developmental testing
  - Many of these diseases associated with severe developmental disabilities

- **How can screening fail?**
  - Home births, delayed screening, religious objections, specimen obtained at wrong time, prematurity
  - FPR (false positive rates) for VLBW, prematurity under 32 weeks,
PAST, PRESENT, FUTURE OF NEWBORN SCREENING

• PKU 1962
• Hypothyroidism 1975
• 4 million babies screened annually
  • 6,000 diagnosed with detectable and treatable diseases
  • Majority of states screen for 29 core conditions (page 69)
  • SCID is recommended to be added
• With new technologies and therapeutic advances, list will grow
• Need to balance legal, ethical, and social concerns
• Prenatal screening
  • First trimester
  • Second trimester
Don't write your name on sand, waves will wash it away.
Don't write your name in sky, wind may blow it away.
Write your name in hearts of people you come in touch with.
That's where it will stay.