Child Hereditary

Nursing 201: Nursing Care of Special Populations

- Overview
- Normal 46 chromosomes
- Errors occur at any time during the process of development
- Genetic research improving methods to control conditions
    - Gene replacement therapy

- Trisomy 21 (Down Syndrome)
- 3 copies of chromosome 21
    - Totaling 47 chromosomes
- Most common chromosomal disorder
- 1: 800 live births
- Increased incidence with:
    - AMA
- Physical Features
    - Small head (microcephaly)
    - Flattened, broad head
    - Mid-face hypoplasia (flattened nasal bridge)
    - Up-slanting eyes
    - Small oral opening with protruding tongue
    - Vertebrae unstable
        - Need clearance
    - Short neck
    - Low set ears
    - Broad chest
    - Possible heart murmurs
    - **R/F congenital heart defects**
    - Short hands that may have single crease
    - Low tone
- Diagnosis
    - Prenatally
        - Amniocentesis
        - Quad screen
    - After birth
        - Based on physical appearance
        - karyotyping
- Nursing considerations
    - Motor development takes twice as long
        - 90% can walk by 3 years old
    - Developmental delays vary
        - Average IQ is 55
        - Extent not present at birth
- Nursing Management
    - Feeding
        - Large tongue
        - Frequent choking
        - Feed slowly
        - PNA risk

- Common Ear Infections
- Joint laxity
- R/F Malignancies
  - Leukemia is 18x greater then general population
- Hair Loss, thinning
- Sleep apnea
  - Suspicion with snoring or unusual sleeping position
- Allergic Dermatitis
- Immune dysfunction
- Need referral to local early intervention services
  - Learn by repetition

- Turner Syndrome
- Females
- Loss of entire X chromosome
- Only known disorder where a fetus can survive despite the loss of an entire chromosome
- Female receives no sex chromosome from sperm of the father, and receive X from mother
- Signs and Symptoms
  - Prematurity
  - Difficulty feeding
  - GERD
  - FTT
  - Lymphedema at birth to hands and feet
- Physical Findings
  - Webbed, thick short neck
  - Short stature
  - Broad chest, wide nipples
  - Nonfunctional ovaries
  - Common heart defects
  - Normal intelligence
    - Visual-perception impairments that predispose to nonverbal learning disabilities
  - Single kidney
- Nursing Management
  - Lack of secondary sexual characteristic by mid-teens
    - Require estrogen therapy for development
    - Remain infertile
  - Endocrine referral
    - Growth hormone treatment (started early in life)
  - Feeding difficulties

- Cri-Du-Chat Syndrome
- “Cat cry” syndrome
  - Distinctive high-pitch, cat-like cry
- Result of a deletion or loss of a portion of chromosome 5
- Often die in infancy
- Physical Findings
  - Microcephaly
  - Round face with widely spaced eyes
  - Low set ears
  - Severe MR
- Nursing Management

- Feeding and poor growth issues
- Family support groups

- Tay Sach's Disease
- Gene level defect
- Lacks Hex A (enzyme necessary for lipid metabolism)
  - Without enzyme, lipid deposits accumulate on nerve cells
  - Cognitive impairment when on brain cells
  - Blindness when deposit on optic nerve
- Incidence primarily in Jewish population
- Signs and Symptoms
  - Appears normal in first few months
    - Except mild hypotonia
  - Around 6 months
    - Loses head control
    - Severe hypotonia
    - Unable to sit up or rollover without support
  - Ophthalmic exam
    - Cherry red macula
    - Caused by lipid deposits
- 1 year old
  - Spasticity (neuro deterioration)
  - Unable to perform simple motor tasks
    - Gag, swallow
- 2 year old
  - Convulsions
  - Blindness
- 3-5 years old
  - Usual life expectancy
  - Ill health, malnutrition, wasting, PNA
  - Tay Sach's Disease
- Diagnosis
  - Amniocentesis (pre-natal)
  - Signs and symptoms on assessment
  - Genetic testing
- Treatment
  - No cure
  - Comfort measures

- Klinefelter's Syndrome
- Presence of one or more X chromosomes
- More common than Turner's Syndrome
- Males
  - XXY chromosome pattern
- Treatment
  - Testosterone to increase male characteristics
- Signs and Symptoms:
  - No distinctive physical characteristics to detect prior to puberty
  - Tall, long legs
  - Small testes
  - Sterile

- Decreased secondary sex characteristics
  - Gynecomastia
- Varying degrees of mental impairment
  - More X's = increased MR

- <u>Phenylketonuria (PKU)</u>
- Disease of metabolism
  - Phenylalanine is an essential amino acid necessary for growth and repair of body cells
  - Results in accumulation of phenylalanine in the blood and brain, causing progressive MR
  - Testing performed in NB period
  - Primary white children
    - 1:10,000-15,000 live births
- Pathophysiology
  - Absence of liver enzyme phenylalanine hydroxylase
  - Prevents conversion of phenylalanine to tyrosine (precursor of epi, thyroxine, melanin)
  - Build up causes severe cognitive impairment
  - Breakdown of phenylalanine (phenylpyruvic acid) spills into urine
    - Musty odor noted
  - Tyrosine
    - Necessary for building body pigment and thyroxine
    - Without it body pigment fades (fair, blonde hair, blue eyes)
    - Fail to meet growth standards
- Signs and Symptoms
  - Seizures
  - Eczema
  - If untreated
    - Mental retardation
      - IQ <20
    - Hypertonicitiy/ spasticity
    - Erratic behavior
      - Head banging
      - screaming
- Diagnosis
  - No detection in utero
    - Operating under mother's enzyme system
  - Heel stick
    - After 2 full days of milk feeding (formula or breast)
    - Check phenylalanine levels
    - Required in all 50 states
- Management
  - Low phenylalanine formula
  - Begin diet early to prevent cognitive impairment
    - Low protein
    - Irreversible once brain injury occurs
    - Difficult to follow indefinitely
    - 20-30 mg phenylalanine per day
    - Low foods
      - OJ, bananas, potatoes, lettuce, spinach, peas
    - High Foods
      - Meat, eggs, milk, aspartame
    - **Blood levels must remain 2-8 mg/dl**

- Genetic testing
  - 50% chance of having a child with PKU
- Prognosis
  - If detected in first few days of life:
    - Diet control to avoid high levels
    - IQ will not be adversely affected
      - Even with adequate control a high percentage of children have some degree of intellectual impairment
  - If not detected:
    - Irreversible brain involvement
  - If pregnant with PKU:
    - Follow low phenylalanine diet for 3 months before conception and during pregnancy

- Galactosemia
- Inborn error of carbohydrate metabolism
- Child deficient in the liver enzyme galactose 1- phosphate uridyl transferase
  - Lactose is broken down into galactose and glucose
  - Galactose is broken down into additional glucose
  - Without the enzyme the second step does not happen
  - Galactose builds up in the bloodstream and spills into the urine
- Rare
- 1: 50,000 births
- Signs and Symptoms
  - Appear normal at birth
  - Symptoms begin abruptly and worsen rapidly
  - After start formula or breast milk
    - Start vomiting and losing weight
  - Hepatic dysfunction
    - Cirrhosis
    - Jaundice
    - Hepatosplenomegaly
  - E. Coli sepsis is common presenting sign
  - Eyes
    - Cataracts (1-2 months)
- Diagnosis
  - NB screening
  - Measure level of enzyme
  - Treatment
  - Untreated
    - Die between 1-4 months
    - Severe cognitive impairment
- Diet
  - Free of galactose
  - No breast feeding
    - Contains lactose
    - Requires lactose free formula
  - Once regulated S&S do not progress
    - If neuro and cataract damage is present, will persist
  - Easier to manage diet then PKU

- Thalassemia
- Group of inherited blood disorders characterized by deficiencies in the rate of production of specific globin chains in Hemoglobin
    - B-thalassemia minor: trait (carrier state)
        - May have mild anemia
    - B-thalassemia major: Cooley's anemia, disease state
        - severe anemia
- Signs and Symptoms
    - Asymptomatic until fetal Hgb has been replaced by adult Hgb (6 months age)
    - Anemia
        - Pallor, irritability, anorexia, Hgb <5, increased serum iron level (no iron in Hgb)
    - Bone marrow
        - Tries to maintain functional level of Hgb
        - Bone marrow hypertrophies in attempt to make more RBC's
        - Bone pain
        - Develop broad base of nose, flattened
        - Down syndrome like eyes
- Signs and Symptoms (continued)
    - Hepatospleenomegaly
        - Excessive iron deposits
        - Scarring of liver
        - Spleen attempt to destroy defective RBC's
        - Abdominal pain, anorexia, vomiting
    - Epistaxis
    - Cardiac murmurs- circulatory overload
        - Arrhythmia, CHF
        - Frequent cause of death
    - Bronzed skin
        - Iron pigmentation
    - Delayed growth and sexual maturation
        - Thalassemia
- Management
    - Prevent heart failure
        - Digoxin, Diuretics, Low Na+ diet
    - Transfuse RBC's Q 2-4 weeks to maintain Hgb 10-12
    - Iron Chelating agent to remove excessive iron stores
        - Desferoxamine Subcutaneous
    - Spleenectomy
    - Bone Marrow Transplant
        - Curative
- Prognosis
    - Most will die during adolescents from cardiac failure

- Severe Combined Immunodeficiency Disorder (SCID)
- Gene level defect
- Sex-linked
- Deficiency of both T and B cells
    - T-cell fights viral, fungal, parasitic infections
- Signs and Symptoms
    - Unable to fight infections
    - Usually in first month of life chronic infection, can't recover completely then re-infect

- Diagnosis
  - Hx of infections
  - Family history
  - Lymphocytopenia
- Treatment
  - Untreated
    - Overwhelming infection and die
  - Germ-free environment
  - Bone marrow transplant
  - Gene therapy
    - Replace child's own cells with those able to produce T-cells
  - Passive immunity with immunoglobin
    - Maintain child in sterile environment
    - Must institute before any infection
    - "bubble boy"

- <u>Muscle Dystrophy</u>
- Group of muscle diseases in childhood
- Lack of protein necessary for muscle contraction
- Progressive weakness and wasting of skeletal muscles
- Types differ in regard to the muscle group affected, progression, inheritance
- most common type is Duchenne MS
- Signs and Symptoms
  - Onset 3-5 years
    - Later than average meeting milestones is indicators
  - Progressive muscle wasting, weakness, possible contractures
  - Calf muscle hypertrophies
  - Waddling gait/ difficulty climbing stairs
    - Loss of ambulation by 9-11 years, then confined to wheelchair
  - Gower's Sign
    - Rise from floor by rolling onto stomach then pushes to knees
    - Presses hands against ankles, knees, thighs (walk up their front) with knees straight
- Gowler's Sign
- Signs and symptoms (continued)
  - Cardiac
    - Muscle weakens/ cardiac enlargement
    - Tachycardia
  - Respiratory
    - Weak and ineffective cough
    - Easily develop PNA
    - Mild MR is common
- Diagnosis
  - Clinical manifestations
  - Muscle biopsy
  - EMG
  - Serum CPK
    - Extremely high in first 2 years before onset of weakness
- Therapeutic Management
  - Remain ambulatory for as long as possible
  - PT

        - Active and passive exercises
    - Splinting
    - Bracing
    - Avoid contractures
- Prognosis
    - Death usually around 20 years
        - Typically r/t to heart or respiratory failure

- Hemophilia
- Group of bleeding disorders in which there is a deficiency of one of the factors necessary for coagulation of the blood
- Blood will eventually coagulate after an injury
- Diagnostics
    - Often noticed after excessive bleeding after a circumcision
    - Bruises heavily from minor injuries
    - Platelet, PT, Thromboplastin all WNL
    - PTT abnormal
- Signs & Symptoms
    - Soft tissue bleeding
    - Painful hemorrhage in joints (hemarthritis)
        - Swollen, warm
    - GI Bleed
    - Frequent epistaxis
- Management
    - Control bleeding
    - Administration of Factor VIII
        - Supplied via whole blood or FFP
        - Prophylactic transfusions may reduce bleeding episodes
    - Prevent injury
        - No contact sports
    - If Injured
    - IV Factor VIII
    - Immobilize injured extremity
    - Ice
    - Avoid sutures
        - Site may bleed
    - NO ASA!
- Hemarthrosis
    - Immobilize the joint (bed rest)
    - Restrict movement
    - Ice
    - Keep joint in alignment