

## Chapter 14

### The Newborn with a Perinatal Injury or Congenital Malformation

- Birth Defects
- Abnormalities that are apparent at birth
- The abnormality may be of
  - Structure
  - Function
  - Metabolism
- May result in a physical or mental disability, may shorten life, or may be fatal
- Classifications of Birth Defects
- Malformations present at birth
- May also be known as *congenital malformations*
- Inborn errors of metabolism
- Disorders of the blood
- Chromosomal abnormalities
- Perinatal injuries
- March of Dimes
- Birth defects cannot be attributed to a single cause.
- Combination of environment and heredity
  - Inherited susceptibility
  - Stage of pregnancy
  - Degree of environmental hazard

- Nervous System
- Neural tube defects
  - Most often caused by failure of neural tube to close at either the cranial or the caudal end of the spinal cord
    - Hydrocephalus
    - Spina bifida
- Hydrocephalus
- Characterized by an increase in CSF within the ventricles of the brain
  - Causes pressure changes in the brain
  - Increase in head size
  - Results from an imbalance between production and absorption of CSF or improper formation of ventricles
- Hydrocephalus (*cont.*)
- Most commonly acquired by
  - An obstruction
  - A sequelae of infection
  - Perinatal hemorrhage
- Symptoms depend on
  - Site of obstruction
  - Age at which it develops
- Hydrocephalus (*cont.*)
- Classifications
  - Noncommunicating

- Obstruction of CSF flow from the ventricles of the brain to the subarachnoid space
- Communicating
  - CSF is not obstructed in the ventricles but is inadequately reabsorbed in the subarachnoid space
- Manifestations of Hydrocephalus
- Depends on time of onset and severity of imbalance
- Classic signs
  - Increase in size of head
  - Cranial sutures separate to accommodate enlarging mass
  - Scalp is shiny
  - Veins are dilated
- Diagnosis and Treatment of Hydrocephalus
- Diagnosis
  - Transillumination
  - Echoencephalography
  - CT scan
  - MRI
  - Ventricular tap or puncture
- Treatment
  - Medications to reduce production of CSF
  - Surgery to place a shunt
- Symptoms of Increasing Intracranial Pressure

- Increased blood pressure
- Decrease in pulse rate
- Decrease in respirations
- High-pitched cry
- Unequal pupil size or response to light
- Bulging fontanel in infants
- Headaches in children due to closed cranial sutures
- Irritability or lethargy
- Vomiting
- Poor feeding
- Ventriculoperitoneal Shunt
- Treatment
  - Medications to reduce CSF production
  - Surgery
  - Shunt acts as a focal spot for infection and may need to be removed if infections persist
- Preoperative and Postoperative Nursing Care
- Pre-Op
  - Frequent head position changes to prevent skin breakdown, head must be supported
  - Head must be supported at all times while being fed
  - Measure head circumference along with other vital signs
- Post-Op
  - Assess for signs of increased intracranial pressure

- Protect from infection
- Depress shunt “pump” as ordered by surgeon
- Position dependent upon multiple factors
- Assess and provide for pain control
- Parent Education
- Teach signs that indicate shunt malfunction may be occurring
  - How to “pump” the shunt
- Signs of shunt malfunction in the older child can include
  - Headache
  - Lethargy
  - Changes in LOC
- Spina Bifida (Myelodysplasia)
- Spina Bifida (Myelodysplasia) (*cont.*)
- Group of CNS disorders characterized by malformation of the spinal cord
- A congenital embryonic neural tube defect with an imperfect closure of the spinal vertebrae
- Two types
  - Occulta (hidden)
  - Cystica (sac or cyst)
- Spina Bifida Occulta
- Minor variation of the disorder
- Opening is small

- No associated protrusion of structures
- Often undetected
  - May have a tuft of hair, dimple, lipoma, or discoloration at the site
- Spina Bifida Occulta (*cont.*)
- Treatment generally not necessary unless neuromuscular symptoms appear, such as
  - Progressive disturbances of gait
    - Foot drop
  - Disturbances of bowel and bladder sphincter function
- Spina Bifida Cystica
- Development of a cystic mass in the midline of the opening in the spine
  - Meningocele
    - Contains portions of the membranes and CSF
    - Size varies
  - Meningomyelocele
    - More serious protrusion of membranes and spinal cord through the opening
    - May have associated paralysis of lower extremities
    - May have poor or no control of bladder or bowel
    - Hydrocephalus is a common complication
- Prevention of Spina Bifida
- Mother takes folic acid 0.4 mg per day prior to becoming pregnant and/or continues to take the folic acid supplement until the 12th week of pregnancy
- Treatment of Spina Bifida
- Surgical closure

- Prognosis is dependent upon extent of spinal cord involvement
- Meningocele
- Contains portions of the membranes and CSF
- If no weakness of the legs or sphincter involvement, surgical correction is performed with excellent results
- Meningomyelocele
- Protrusion of the membranes and spinal cord through the opening
- Surgical intervention is done for cosmetic reasons and to help prevent infection
- *Habilitation* is usually necessary post-op because the legs remain paralyzed and the patient is incontinent of urine and feces
- Habilitation
- Patient is disabled from birth
- Aim is to minimize the child's disability
- Constructively use all unaffected parts of the body
- Every effort is made to help the child develop a healthy personality so that he or she may experience a happy and productive life
- Nursing Care of Spina Bifida
- Prevent infection of, or injury to, the sac
- Correct positioning to prevent pressure on sac
- Prevent development of contractures
- Good skin care
- Adequate nutrition
- Accurate observations and charting

- Education of the parents
- Continued medical supervision and habilitation
- Nursing Care of Spina Bifida (*cont.*)
- Upon delivery, the newborn is placed in an incubator
- Moist, sterile dressing of saline or an antibiotic solution may be ordered to prevent drying of the sac
- Protection from injury and maintenance of a sterile environment for the open lesion are essential
- Nursing Care of Spina Bifida (*cont.*)
- Size and area of sac are checked for any tears or leakage
- Extremities are observed for deformities and movement
- Head circumference is measured
- Fontanelles are observed to provide a baseline for future assessments
- Nursing Care of Spina Bifida (*cont.*)
- Complications that can be life-threatening must be monitored
  - Meningitis
  - Pneumonia
  - UTI
- Urological monitoring
- Skin care
- Feeding
- Potential for latex allergy



- Gastrointestinal System
- Cleft Lip
- Characterized by a fissure or opening in the upper lip
- Failure of maxillary and median nasal processes to unite during embryonic development
- Many cases are hereditary, others are environmental
- Appears to occur more often in boys than girls
- Treatment
- Initial repair of cleft lip is known as *cheiloplasty*
- Repair by 3 months of age
- Infant may have to have elbow restraints to prevent the infant from scratching the lip
- A special syringe or bottle may be needed to assist in feeding the child until surgery has occurred
- Postoperative Nursing Care
- Prevent infant from sucking and crying
- Careful positioning to avoid injury to operative site
- Preventing infection and scarring by gentle cleansing of suture lines to prevent crusts from forming
- Providing for the infant's emotional needs by cuddling and other forms of affection
- Providing appropriate pain relief measures
- Feeding
- Fed by medicine dropper until wound is completely healed (about 1 to 2 weeks)
- Cleanse the mouth by giving the infant small amounts of sterile water at the end of each feeding session

- Cleft Palate
- The failure of the hard palates to fuse at the midline during the 7th to 12th weeks of gestation
- Forms a passageway between the nasopharynx and the nose
  - Increases risk of infections of the respiratory tract and middle ears
- Cleft Palate Treatment
- Goals of therapy
  - Union of the cleft
  - Improved feeding
  - Improved speech
  - Improved dental development
  - The nurturing of a positive self-image
- Multidisciplinary Team Care
- Along with the patient and family
  - Psychologist
  - Speech therapist
  - Pediatric dentist
  - Orthodontist
  - Social worker
  - Pediatrician
- Other Factors
- Psychosocial adjustment of the family

- Follow-up care
- Home care
- Surgery between 1 year and 18 months of age
- Postoperative Treatment and Nursing Care
- Nutrition
  - Diet is progressively advanced
  - No food through straws to prevent sucking
- Oral hygiene
  - Follow each feeding with clear water to cleanse the mouth
- Speech
  - Encourage children to pronounce words correctly
- Diversion
  - Crying should be avoided whenever possible; play should be of the quiet type (e.g., coloring, drawing, reading to the child)
- Complications
  - Ear infections and tooth decay
- Musculoskeletal System
- Clubfoot
- Most common deformities
- Congenital anomaly
  - Foot twists inward or outward
- *Talipes equinovarus* is the most common type
  - Feet turned inward
  - Child walks on toes and outer borders of feet

— Generally involves both feet

- Clubfoot (*cont.*)
- Treatment and Nursing Care
- Started as soon as possible or bones and muscles will continue to develop in an abnormal manner
- Conservative treatment
  - Splinting or casting to hold foot in correct position
  - Passive stretching exercises
- If not effective after about 3 months, surgical intervention may be indicated
- Parent Education
- Stress importance of complying with physician orders to prevent skin breakdown and possible isolation of the older child
- The nurse should review with the parents
  - Cast care
  - Emotional support
- Developmental Hip Dysplasia
- Developmental Hip Dysplasia (*cont.*)
- *Hip dysplasia* applies to various degrees of deformities, subluxation or dislocation (can be partial or complete)
- Head of femur is partly or completely displaced
- Seven times more common in girls
- More apparent as infant/toddler begins walking

- Developmental Hip Dysplasia (*cont.*)
- Usually discovered at routine health checks during the first or second month of life
- Most reliable sign is limited abduction of the leg on the affected side
- Diagnostics for Hip Dysplasia
- *Barlow's test*: upon adduction and extension of the hips (with health care provider providing stabilization to the pelvis), may "feel" the dislocation actually occur
- *Ortolani's sign* (or click): health care provider can actually feel and hear the femoral head slip back into the acetabulum under gentle pressure
- Treatment of Hip Dysplasia
- Hips are maintained in constant flexion and abduction for 4 to 8 weeks
  - Keeps head of femur within the hip socket
- Constant pressure enlarges and deepens acetabulum
- Can use a *Pavlik harness* to provide the necessary positioning
- Surgical intervention may be necessary
- Pavlik Harness, Body Cast, and Traction
- Nursing Care of Infant/Child in a Spica Cast
- Neurovascular assessment of affected extremities
- Place firm, plastic-covered pillows beneath the curves of the cast for support
- In the older child, a "fracture" bedpan should be readily available for toileting
- Head of bed slightly elevated to help drain any body fluids away from cast
- Frequent changes of position are needed to prevent skin breakdown
- Nursing Care of Infant/Child in a Spica Cast (*cont.*)

- Toys that are small enough to “hide” in the cast should not be given to the child
- Important to meet everyday needs
- A special wagon with pillows inside it for support is one of the safest ways to transport a child in a spica cast
- Metabolic Defects
- Inborn errors of metabolism involve a genetic defect that may not be apparent until after birth
- Symptoms to report would include
  - Lethargy
  - Poor feeding
  - Hypotonia
  - Unique odor to body or urine
  - Tachypnea
  - Vomiting
- Phenylketonuria (PKU)
- Faulty metabolism of phenylalanine, an amino acid essential to life and found in all protein foods
- Infant unable to digest this essential acid and phenylalanine accumulates in blood and is found in the urine within the first week of life
- Results in severe mental retardation if not caught early
- Phenylketonuria (PKU) (*cont.*)
- Appears normal at birth
- By the time urine test is positive, brain damage has already occurred
- Delayed development apparent at 4-6 months
- May have failure to thrive, eczema, or other skin conditions

- Child has a musty odor
- Personality disorder
- Occurs mainly in blonde, blue-eyed children
- Results from a lack of tyrosine (needed for melanin formation)
- PKU Diagnostics
- *Guthrie* test
- Blood for this test should be obtained 48 to 72 hours after birth
- Preferably after the infant has ingested proteins
- Many states require this test to be performed prior to discharge from hospital
- PKU Treatment
- Close dietary management
- Frequent evaluation of blood phenylalanine level
- Synthetic food that provides enough protein for growth and tissue repair
  - Special formulas are available
    - Infants: Lofenalac or Phenex-1
    - Children: Phenyl-free
    - Adolescents: Phenex-2
- PKU Nursing Care
- Teach parents importance of reading food labels
- Following up as required with health care provider for blood tests
- Referral to a dietitian is helpful in providing parental guidance and support
- Genetic counseling may also be indicated

- Health Promotion
- Children with PKU must avoid the sweetener aspartame (NutraSweet) because it is converted to phenylalanine in the body
- Maple Syrup Urine Disease
- Defect in the metabolism of branched-chain amino acids
- Causes marked serum elevations of leucine, isoleucine, and valine
- Results in acidosis, cerebral degeneration, and death within 2 weeks if not treated
- Maple Syrup Urine Disease (*cont.*)
- Appears healthy at birth, but problems soon develop
- Feeding difficulties
- Loss of the Moro reflex
- Hypotonia
- Irregular respirations
- Convulsions
- Maple Syrup Urine Disease (*cont.*)
- Manifestations
  - Urine, sweat, and cerumen (earwax) have a characteristic maple syrup odor caused by ketoacidosis
  - Diagnosis confirmed by blood and urine tests
- Maple Syrup Urine Disease Treatment and Nursing Care
- Treatment
  - Removing the amino acids and their metabolites from the body tissues



- Hydration and peritoneal dialysis to decrease serum levels
- Lifelong diet low in amino acids leucine, isoleucine, and valine
- Exacerbations are usually related to degree of abnormality of leucine level
- Infection can be life-threatening
- Galactosemia
- Unable to use galactose and lactose
  - Enzyme needed to help the liver convert galactose to glucose is defective or missing
  - Results in an increased serum galactose level (galactosemia) and in the urine (galactosuria)
- If untreated can cause
  - Cirrhosis of the liver
  - Cataracts
  - Mental retardation
- Galactose is present in milk in the form of sugar; therefore, early diagnosis is essential
- Galactosemia (*cont.*)
- Begins abruptly, worsens gradually
- Early signs
  - Lethargy
  - Vomiting
  - Hypotonia
  - Diarrhea
  - Failure to thrive
- Symptoms begin as the newborn is fed
- Jaundice may be present

- Galactosemia Treatment and Nursing Care
- Milk and lactose-containing products are eliminated from the diet
- Breastfeeding *must* be stopped
- Lactose-free formulas or soy protein-based formulas are often used instead
- Parental support and education is essential
- Chromosomal Abnormalities
- Down Syndrome
- Most common chromosomal abnormality
- Risk increases with
  - Mothers 35 years and older
  - Fathers 55 years and older
- Infant has mild to severe mental retardation
- Some physical abnormalities are also seen
- Down Syndrome (*cont.*)
- Three phenotypes
  - Trisomy 21
    - Most common
    - There are three number 21 chromosomes instead of the usual two
    - Results from nondisjunction (failure to separate)
  - Mosaicism
    - Occurs when both normal and abnormal cells are present
    - Tend to be less severely affected in appearance and intelligence
  - Translocation of a chromosome

- A piece of chromosome in pair 21 breaks away and attaches itself to another chromosome
- Down Syndrome (*cont.*)
- Screening for this is offered during prenatal care starting around week 15 of gestation
  - Allows parents the opportunity to decide on whether to continue or terminate the pregnancy
- “Quad Test”: Alpha-fetoprotein (AFP), hCG, unconjugated estriol, inhibin-A levels are used for diagnosis
- Amniocentesis is most accurate
- Down Syndrome Manifestations
- Down Syndrome Manifestations (*cont.*)
- Limp, flaccid posture caused by hypotonicity of muscles
  - More difficult to position and hold
  - Contributes to heat loss
- Prone to respiratory illnesses and constipation due to the hypotonicity
- Incidence of acute leukemia is higher
- Alzheimer’s disease more common to those who reach middle adult life
- Encourage parents to express their feelings and concerns
- Provide parents with support and community referrals
- Developmental Milestones
- Sitting
- Rolling over
- Sitting alone
- Crawling

- Creeping
- Standing
- Walking
- Talking
- Self-Help Skills
- Eating
- Toilet training
- Dressing
- Perinatal Injuries
- Hemolytic Disease of the Newborn (Erythroblastosis Fetalis)
- Becomes apparent in utero or soon after birth
- Rh-negative mother and Rh-positive father produce Rh-positive fetus
- Even though maternal and fetal blood do not mix during pregnancy, small leaks may allow fetal blood to enter the maternal circulation causing the mother's body to start producing antibodies that cross the placenta and destroy the blood cells of the fetus, which can cause anemia and heart failure in the developing/growing fetus
- Erythroblastosis Fetalis:  
Maternal Sensitization
- Erythroblastosis Fetalis:  
Maternal Sensitization (*cont.*)
- Mother accumulates antibodies with each pregnancy
- Chance of complications occurs with each subsequent pregnancy
- Severe form, *hydrops fetalis*, progressive hemolysis causes anemia, heart failure, fetal hypoxia, and anasarca

- Erythroblastosis Fetalis  
Diagnosis and Prevention
- Maternal health history that includes
  - Previous Rh sensitizations
  - Ectopic pregnancy
  - Abortion
  - Blood transfusions
  - Child who developed jaundice or anemia during a neonatal period
- Indirect Coombs' test will indicate previous exposure to Rh-positive antigens
- Erythroblastosis Fetalis  
Diagnosis and Prevention (*cont.*)
- Confirmed by amniocentesis and monitoring of bilirubin levels in the amniotic fluid
- Fetal Rh status can be determined non-invasively via free DNA in maternal plasma
- Diagnostic studies will help the physician to determine if early interventions, such as induction of labor or intrauterine fetal transfusions, are needed
- Erythroblastosis Fetalis  
Diagnosis and Prevention (*cont.*)
- Use of Rh(D) immune globulin (RhoGAM)
- Administered within 72 hours of delivery with an infant who is Rh-positive, an ectopic pregnancy, or after an abortion
- May also be given to the pregnant woman at 28 weeks gestation
- Erythroblastosis Fetalis  
Manifestations
- Direct Coombs' test on umbilical cord blood
- Symptoms vary

- Anemia caused by hemolysis of large numbers of erythrocytes
- *Pathological jaundice* occurs within 24 hours of delivery; liver cannot handle the amount of hemolysis, bilirubin levels rise rapidly
- Enlargement and edema of liver and spleen
- Oxygen-carrying capacity of the blood is diminished, including blood volume
- Infant at major risk of shock or heart failure
- Erythroblastosis Fetalis Manifestations (*cont.*)
- Kernicterus—bilirubin has reached toxic levels
- Accumulated bilirubin in the brain tissue can cause serious brain damage and permanent disability
- Infant will have jaundice along with
  - Irritability
  - Lethargy
  - Poor feeding
  - High-pitched, shrill cry
  - Muscle weakness
  - Progresses to opisthotonos
  - Seizures
- Erythroblastosis Fetalis Treatment
- Prompt identification
- Laboratory tests
- Drug therapy
- Phototherapy
- Exchange transfusions, if indicated
- Erythroblastosis Fetalis Nursing Care
- Ensure eyes are protected from phototherapy

- Cover gonads
- Provide incubator care
- Central line care (usually the umbilical vein)
- Observe newborn's color
- Apply wet, sterile compresses to the umbilicus, if ordered, until transfusions are complete
- Nursing Tip
- Assessing jaundice involves
  - The skin and the whites of the eyes assume a yellow-orange cast
  - Blanching the skin over bony prominences enhances the evaluation of jaundice
- Jaundice that occurs on the first day of life is always pathological and requires prompt intervention
- Home Phototherapy
  - Used for newborns with mild to moderate physiological (normal) jaundice
  - Less costly
  - May decrease the need for hospitalization
- Intracranial Hemorrhage
  - Most common type of birth injury
  - May result from trauma or anoxia
  - Occurs more often in preterm infants
  - May also occur during precipitate delivery or prolonged labor
  - Signs and symptoms vary depending on severity

- Intracranial Hemorrhage (*cont.*)
- Diagnosis
  - History of traumatic delivery
  - CT or MRI scan
  - Evidence of an increase in CSF pressure
- Treatment
  - Oxygen
  - Gentle handling
  - Elevated head
  - Medications may be prescribed
  - Care with feeding because sucking reflex may be affected
- Intracranial Hemorrhage (*cont.*)
- If convulsion occurs, notify physician immediately
- Be ready to answer the following questions
  - Were the arms, legs, or face involved?
  - Was the right or left side of the body involved?
  - Was the convulsion mild or severe?
  - How long did it last?
  - What was condition of infant before and after the seizure (i.e., vital signs, skin color)?
- Transient Tachypnea of the Newborn (TTN)
- Characterized by
  - Tachypnea



- May also include
  - Chest retractions
  - Grunting
  - Mild cyanosis
- Often referred to as *respiratory distress syndrome*, type II
- Typically resolves suddenly after 3 days
  - May be caused by slow absorption of fluid in lungs after birth
- Supportive nursing and medical care
- Meconium Aspiration Syndrome
- In utero
  - Fetus expels meconium into amniotic fluid
  - Cord compression or other condition interrupts fetal circulation
  - If asphyxia or acidosis occurs, fetus may have gasping movements that cause meconium-stained amniotic fluid to be drawn into the lungs
- At delivery
  - Can occur if newborn inhales before nose and mouth have been suctioned
- Meconium Aspiration Syndrome (*cont.*)
- Symptoms
  - Respiratory distress
  - Nasal flaring
  - Retractions
  - Cyanosis
  - Grunting

- Rales and rhonchi
- Tachypnea may persist for several weeks
- Treatment
  - Warmth
  - Oxygen
  - Supportive care
  - NICU
- Neonatal Abstinence Syndrome (NAS)
- Caused by fetal exposure to drugs in utero
- Many illicit drugs cross the placental barrier; therefore, an infant born to a woman who is an addict will suffer drug withdrawal after birth
- Infant may also have long-term developmental and neurological deficits
- Neonatal Abstinence Syndrome (NAS) (*cont.*)
- Symptoms
  - Body tremors and hyperirritability (primary sign)
  - Wakefulness
  - Diarrhea
  - Poor feeding
  - Sneezing
  - Yawning
- Testing
  - Meconium may be more accurate than neonatal urine testing for presence of drugs

- Treatment
  - Swaddling
  - Quiet environment
  - Observe for seizures
  - Phenobarbital
- Infant of Diabetic Mother
- Large amounts of glucose are transferred to fetus
- Causes fetus to become hyperglycemic
- Fetal pancreas produces large amount of fetal insulin
- Leads to hyperinsulinism, along with excess production of protein and fatty acids, often results in an LGA newborn weighing 9 pounds (4082 g) or more (macrosomia)
- Infant of Diabetic Mother (*cont.*)
- After delivery, infant may have low blood glucose levels and Cushingoid appearance or look healthy
- May have developmental deficits and suffer complications of RDS
- Suffers from
  - Hypoglycemia
  - Hypocalcemia
  - Hyperbilirubinemia
- Infant of Diabetic Mother (*cont.*)
- Monitor
  - Glucose levels

- Vital signs
- Signs of irritability
- Tremors
- Respiratory distress

- Glucose levels below 40 mg/dL can result in rapid and permanent brain damage
- Question for Review
- How can prenatal care prevent neural tube defects?