Spina Bifida is a developmental abnormality due to insufficient closure of the neural tube by the 28th day of gestation. This defect usually occurs in low thoracic, lumbar, or sacral regions and affects the central nervous, musculoskeletal, and urinary systems.

Spina Bifida

- A single etiology has not been identified
- However, causative factors include
  - Genetic predisposition
  - Environmental influence
  - Low levels of maternal folic acid
  - Maternal hyperthermia
  - Certain drugs
Spina bifida (cont’d)

Types of Spina Bifida

1. **Spina Bifida Occulta**
   An impairment & non-fusion of the spinous processes of a vertebrae, however, the spinal cord remains intact. There is usually no disability.

2. **Spina Bifida Cystica**
   presents with a cyst-like protrusion through the non-fused vertebrae, which results in impairment

Spina Bifida

- **Forms of Spina Bifida Cystica**
  - **Meningocele**: herniation of meninges and CSF into a sac that protrudes through the vertebral defect. The spinal cord remains within the canal.
  - **Myelomeningocele**: a severe form characterized by herniation of meninges, CSF, and the spinal cord extending through the defect in the vertebrae. The cyst may or may not be covered by the skin.

Physical Therapy

- Family education regarding positioning, handling, ROM & TE.
- ROM
- Facilitate developmental milestones
- Skin care
- Strengthening
- Balance & mobility
- Adaptive equipment
Cerebral Palsy

- CP is an umbrella term used to describe movement disorders due to brain damage that are
  - Non-progressive
  - Acquired in utero, during birth, or infancy

- The brain damage decreases the brain's ability to monitor & control nerve & voluntary muscle activity

Cerebral Palsy

- **Causative Factors**
  - Lack of oxygen
  - Maternal infections
  - Drug or alcohol abuse
  - Placental abnormalities
  - Toxemia
  - Prolonged labor
  - Prematurity
  - Rh incompatibility
  - Meningitis, CVA, seizures, head injury

Cerebral Palsy

- **Characteristics**
  - Vary from mild and undetectable to severe loss of control accompanied by profound mental challenge
  - All types of CP demonstrate abnormal muscle tone, impaired movement, presence of abnormal reflexes & impaired mobility
Cerebral Palsy

- **Primary Motor Patterns**
  - Spastic
  - Athetoid

- **Distribution of Involvement**
  - Monoplegia
  - Diplegia
  - Hemiplegia
  - Quadriplegia

Treatment CP

- Lifelong process
- Family & caregiver education
- Normalization of tone
- Stretching, strengthening
- Motor learning & developmental milestones
- Positioning, WBing activities, mobility skills
- Splinting, assistive devices, special seating

Genetic Disorders

- The abnormality or “injury” occurs at the level of the gene. Affects one or more chromosomes during gestation.
  1. Down Syndrome
  2. Duchenne Muscular Dystrophy (DMD)
  3. Cystic fibrosis
  4. Osteogenesis imperfecta (OI)
**Down syndrome**
- Leading chromosomal cause of mental challenges
- Extra 21st chromosome (Trisomy 21)
- Characteristics
  - Hypotonicity
  - Joint hypermobility
  - Upwardly slanting epicanthal folds (the skinfold of the upper eyelid covering the inner corner of the eye)
  - Flat nasal bridge
  - Pes planus
  - T-L scoliosis
  - Patellar instability

**Down Syndrome**
- Motor development is slow
- Ligamentous laxity 2° collagen defect

**INTERVENTION**
- Emphasize exercise & fitness
- Stability
- Maximizing respiratory function
- Education for caregivers

**Duchenne Muscular Dystrophy**
- A progressive disorder
- Caused by absence of the gene required to produce the muscle proteins dystrophin & nebulin
- Without these proteins, cell membranes weaken, myofibrils are destroyed, and muscle contractility is lost
- Fat & connective tissue eventually replace muscle
- Death occurs from cardiopulmonary failure prior to age 25, but usually in the teenage years
Duchenne Muscular Dystrophy

- Caused by an X-linked recessive trait (the mother is a silent carrier and only male offspring will manifest the disease)
- Characteristics usually start to show between 2 & 5 years of age: progressive weakness, disinterest in running, falling, toe walking, excessive lordosis, pseudohypertrophy of muscle,
- progressive impairments with ADLs & mobility starts at age 5 and ends with the inability to ambulate

Treatment
- Family and caregiver education
- Respiratory function
- Submaximal exercise
- Mobility skills
- Adaptive equipment

http://www.youtube.com/watch?v=bI6utCce_3g

Osteogenesis Imperfecta

- Autosomal dominant disorder of collagen synthesis that affects bone metabolism
- "brittle bones"
- Handling training is imperative.
- Structured environment
- Frequency of fracture decreases after puberty

Same goals and interventions: ROM, strengthening, functional activities and gait training, orthotic recommendations as appropriate.
Questions???