Spina Bifida Update 2011

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Objectives

- Know the proposed etiology of Spina Bifida
- Know the importance of folic acid in the prevention of Neural Tube Defects
- Be familiar with prenatal diagnosis and management options for Spina Bifida
- Know the signs, symptoms, and treatment of primary conditions commonly associated with MMC
- Be familiar with secondary conditions associated with MMC
- Be familiar with recent research trials in the treatment of spina bifida and its sequelae
Lecture Outline

- Prevalence, incidence, etiology and prevention of spina bifida
- Prenatal diagnosis and management
- Types of spina bifida
- Motor effects
- Sensory effects
- Hydrocephalus and Arnold-Chiari II
- Spine abnormalities
- Neurogenic bowel and bladder
- Secondary conditions
- New Approaches to care
Prevalence

- 2nd most common physical disability in childhood
- 1/1000 live births nationally
- (1-2/1000 live births in North Carolina)
Incidence

- *Increased risk in those of Irish, German, or Hispanic descent*
- *Decreased risk in Asians and Pacific Islanders*

*Duke Center for Human Genetics Study*

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Etiology

- Polygenic Inheritance
- Environmental Influences (Nutrition, heat, valproic acid, Agent Orange, pesticide exposure)
- MTHFR
- Folic Acid
Prenatal Diagnosis

- Alpha-fetoprotein in amniotic fluid
- Acetylcholinesterase in amniotic fluid
- Fetal Ultrasound
Prenatal Options

- **Termination of pregnancy**
- **C-section delivery**
- **Fetal surgery**
Spina Bifida Occulta

- 1 of 5 people
- Failure of vertebrae to fuse (L-S level)
- Associated spinal cord or nerve root malformations
- Pigmented nevus, angioma, tuft of hair, dimple or dermal sinus
- KEY RISK = tethered cord
- Not usually associated with Arnold-Chiari malformation
Spina Bifida Cystica

- **Meningocele** = meningeal cyst filled with fluid (neuro exam may be normal)
  - Not associated with hydrocephalus

- **Myelomeningocele** = sac also contains dysplastic nerve tissue
  - ✓ (with A/C malformation + Hydrocephalus)
Nerve Involvement

- NOT ALWAYS SYMMETRIC
- Increased or decreased muscle tone
- Muscle weakness
- Decreased sensation
- Neurogenic Bowel
- Neurogenic Bladder
- Vasomotor dysfunction
Motor Levels

- Thoracic
- L1-L2
- L3
- L4
- L5
- S1
- S2-S5
Sensory Levels

- T4
- T10
- L1
- L2
- L3
- L4
- L5
- S1
- S2
- S3-S5
Hydrocephalus

- Most commonly due to obstruction of CSF flow
- VP Shunt done in 85-90% of MMC
  (1/3 will require revision at some time)
Hydrocephalus

- **Symptoms:** Headache, irritability, N/V

- **Acute Signs:** Large head, bulging fontanelle, prominent scalp veins, somnolence

- **Chronic Signs:** Decreased school performance, personality changes, decreased fine motor skills
Arnold-Chiari II Malformation

- **Definition**: Medulla, Pons, 4th ventricle +/- cerebellar vermis herniated into the cervical spinal canal
- **Incidence**: 80-90% of those with MMC
- **Symptomatic**: ~20%
- **Is it pushed (hydrocephalus), pulled (tethered cord), or crowded out?**
Arnold-Chiari II Malformation

- Stridor
- Laryngeal nerve palsy / vocal cord paralysis
- Periodic breathing
- Sleep apnea
- Dysphagia
- Aspiration pneumonia
- CENTRAL RESPIRATORY DYSFUNCTION
  (now the most common cause of death in MMC)
Spine Abnormalities

- Bifid Spine
- Vertebral fusion
- Hemivertebrae
Neurogenic Bladder
Neurogenic Bladder

- Incontinence
- UTI
- Hydronephrosis
- Pyelonephritis
- Renal Calculi

- INTERMITTENT CATHETERIZATION!
  (Only 5% will void with continence)
Bladder Care

- Intermittent catheterization
- Mitrafanoff Procedure
- Hyaluronic acid injections to reduce reflux
- Botulinum toxin injections to reduce bladder/sphincter pressures *(off-label)*
- Neuromodulation using transcutaneous approaches with interferential electrostimulation *(Experimental)*
- Nerve re-routing for “re-innervation” *(XIAO procedure)* *(Experimental)*
Neurogenic Bowel

- Colon, Rectum and internal anal sphincter are affected
- Decreased motility
- Constipation
- Fecal overflow
- Incompetent Rectum
- NEED BOWEL TRAINING PROGRAM!
Neurogenic Bowel

- High fiber diet
- Stool softeners
- Gastro colic reflex
- Suppositories
- Enemas
- Biofeedback (if intact anocutaneous reflex)
- (M)ACE procedure
MACE Procedure
Secondary Conditions

- Fractures
- Hypertension
- Hip dislocation
- Scoliosis
- Kyphosis
- Foot anomalies
- Pressure ulcers
- Burns
- Obesity
- Precocious Puberty
- UTI’s
- Hydronephrosis
- Latex Allergy
- Syringomyelia
- Tethered Cord
- Rotator Cuff tears
- CTS
- Ulnar Neuropathy
Team Approach

- PM&R
- ORTHOPEDICS
- NEUROSURGERY
- UROLOGY
- PT
- OT
- MSW
- EDUCATOR
Crucial Periods

- After diagnosis
- After Birth
- First Year of Life
- Preschool
- 1st grade
- Middle School
- High School
- Transition to Adulthood
Fetal Surgery

- Reversal of the hindbrain herniation (the Chiari II malformation)
- Reduced Need for VP shunt
- *Reduced Incidence or severity of motor impairment
Susan (& Anna)
Questions ?