Mysteries of the Myelomeningocele Patient: A Case Series of Neurosurgical Challenges

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Learning Objectives
1. PNPs will understand the pathophysiology of myelomeningocele and how neurologic function is impacted
2. PNPs will become familiar with the surgical options available to myelomeningocele patients across the life span
3. PNPs will appreciate the changing psychosocial needs of the myelomeningocele patient population over time
4. PNPs will recognize the importance of a strong multidisciplinary team for the myelomeningocele patient population

Spina Bifida
• Latin for “split spine”
• Global term used to describe a group of neural tube defects
  – Closed defect
  – Meningocele = meninges protrude through spinal defect, but spinal cord is normal
  – Myelomeningocele = most severe, spinal cord and meninges both damaged and protrude through the spinal defect

Myelomeningocele
• A defect of primary neurulation
  – Failure of fusion of the caudal region of the neural tube
  – Primary neurulation is completed in the first month of pregnancy
  – Pathophysiology: during neurulation
• In the U.S., incidence is 1500 babies born per year
  – Highest rates in Hispanic women (3.8 per 10,000 live births)
  – Non-Hispanic black or African-American: 2.73 per 10,000 live births
  – Non-Hispanic white: 3.09 per 10,000 live births

Maternal Risk Factors
• Family history of previous neural tube defect
• Alcohol consumption
• Obesity
• Maternal diabetes mellitus
• Fever
• **Folic acid deficiency or altered folate metabolism
  – In 1992, US Public Health Service recommended that women of childbearing age consume 400 mcg folic acid
  – In 1998, US Food & Drug Administration mandated that “enriched” grain products (breads, cereals, rice) be supplemented with folic acid
  • Since 1998, US has seen a 31% decrease in the prevalence of neural tube defects
**Diagnosis**

- During Pregnancy
  - Alpha-fetoprotein (AFP): high serum levels can indicate a neural tube defect
  - Ultrasound: examination of the spine
  - Amniocentesis: high levels of AFP in the amniotic fluid can indicate a neural tube defect
  - Prenatal MRI: further characterize brain anomalies associated with MM
- After Pregnancy
  - Neural tube defect may not be diagnosed until after birth if the mother did not receive prenatal care, or if ultrasounds were inconclusive

**Neurologic Function**

- Functional abilities of the child correspond with the anatomical level of the lesion
- Functional abilities can be better or worse than expected based on the anatomical level
  - Worse function than expected in 48% of patients
  - Improved function than expected in 14% of patients

**Case 1: HH**

- Neural tube defect diagnosed prenatally via anatomic ultrasound & elevated AFP levels
- Born at 37 6/7 gestation via urgent C-section due to maternal hypertension

<table>
<thead>
<tr>
<th>Transferred to</th>
<th>Surgical repair of myelomeningocele</th>
<th>Discharged home</th>
<th>VP shunt placed</th>
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<tbody>
<tr>
<td>DAY 0</td>
<td>DAY 1</td>
<td>DAY 8</td>
<td>DAY 14</td>
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**Follow up Visit**

- **RESPIRATORY DISTRESS**  
  (https://www.youtube.com/watch?v=oeoAze-chng)
- What is your differential?

**Surgical Interventions**

- VP shunt revision (1 mo, 20 days)
- VP shunt revision (8 mo, 15 days)
- VP shunt revision (13 mo)
- VP shunt revision (14 mo)
- Tracheostomy placement (3 mo, 20 days)
- VP shunt revision (8 mo, 12 days)
- VP shunt revision (8 mo, 10 days)
- VP shunt revision (6 mo, 10 days)
- VP shunt revision (3 mo, 10 days)
- VP shunt revision (6 mo, 10 days)
Myelomeningocele Repair: Options

- Standard postnatal repair
- Intrauterine repair
- Hydrocephalus watch

Standard Postnatal Repair

- Spinal defect is protected immediately after birth
- Taken to OR within 24-48 hours of birth for surgical repair
- Neurosurgeon repairs the dura around the spinal cord
- Plastic surgeon may help to close the muscles & skin around the defect
- Goals of surgery
  - Prevent infection
  - Maintain current neurologic function

Intrauterine Repair

- First performed in 1994 via endoscopic repair – unsuccessful
- In 1997, 2 institutions began via incision into the uterus – promising results in improvement of Chiari II malformations, decreased need for shunting for hydrocephalus
- Uterus is exposed, hysterotomy performed, intrauterine volume maintained by fluids, surgery is performed via the hysterotomy opening, neural tube defect is repaired
- Mother is kept on bed rest
- Infants delivered at 36 weeks gestation via C-section

MOMs Study

- Conducted at 3 institutions, 2003-2010
- 183 eligible women were randomized
- In December 2010, the trial was terminated early due to the determined efficacy of prenatal surgery
- Outcomes:
  - Pregnancy complications
  - Need for shunting
  - Evidence of hindbrain herniation (Chiari malformation)
  - Score on the Bayley Mental Development Index
  - Functional abilities vs. anatomical level of the lesion

Outcomes

- Average gestational age at birth
  - Prenatal surgery group: 34.1 weeks
  - Postnatal surgery group: 37.3 weeks
- Need for shunting
  - Prenatal surgery group: 68%
  - Postnatal surgery group: 98%
- No evidence of hindbrain herniation
  - Prenatal surgery group: 30%
  - Postnatal surgery group: 4%
- Rates of “moderate or severe” hindbrain herniation
  - Prenatal surgery group: 25%
  - Postnatal surgery group: 67%
- Syringomyelia
  - Prenatal surgery group: 39%
  - Postnatal surgery group: 58%
- Chiari decompression surgery
  - Prenatal surgery group: 1%
  - Postnatal surgery group: 5%

- 2 perinatal deaths in each group
- Pregnancy complications more common in the prenatal-surgery group
- Motor & mental function improved in the prenatal-surgery group

Post-MOMs Study

- 8-10 centers now offer intra-uterine repair
- Modifications & improvements in surgical technique have cut rates of maternal blood loss intraoperatively
- Intrauterine fetal distress & postnatal complications are still risks
- Following intrauterine repair, if mothers are stable, they are allowed to return home for delivery
- Centers are examining inclusion criteria
Hydrocephalus Watch

- 40-90% of babies develop hydrocephalus
- Manage inpatient
  - Monitor head circumference, serial ultrasounds, clinical symptoms
- Manage outpatient
  - Monitor head circumference, serial scans (CT vs. MRI), clinical symptoms
- Need for CSF diversion
  - Endoscopic third ventriculostomy
  - VP shunt placement

Case 2: MB

- Myelomeningocele diagnosed prenatally
- To OR on date of birth for closure of myelomeningocele & placement of VP shunt
- Social History:
  - Biological mother with long history of mental health illness, was admitted to psychiatric hospital during MB's first year of life
  - Placed in the care of her maternal aunt, suffered from severe neglect, resulting in developmental delay
  - Transferred to foster care around age 2, was later adopted by this family and slowly caught up to developmental milestones

MB Years 0-16

- Received therapies and caught up to developmental milestones
- Followed annually in Spina Bifida clinic (Neurodevelopment, Neurosurgery, Urology, Orthopedics)
- Required 1 revision of VP shunt at age 15 due to disconnected distal tubing

MB Year 16-17

- Leg & back pain → concern for tethered cord → spinal cord detethering surgery
- Continued leg pain → headaches → admitted for VP shunt revision
- Frequent “episodes” with altered mental status & bradycardia
- Loss of lower extremity, bowel, & bladder function
- What is your differential?

MB: 3 month hospitalization

- Work up
  - Labs, imaging
  - Consults: Rehab, Neurology, Cardiology, Psychiatry, Rheumatology, Ophthalmology, Medically Complex Team, Child Life
- Surgical interventions over 2 months:
  1. Shunt revision x 2
  2. Shunt externalized & re-internalized
  3. Posterior fossa decompression
  4. 3rd Shunt revision
  5. Bitemporal cranial vault expansion
- 1 month Rehab stay

MB: Follow Up

- Continued seizures/events
- Plan of care developed to allow MB to return to school
- Subsequent hospitalizations for UTIs
- Most recent surgery for suprapubic catheter placement to allow more independence
- Continued follow up by Pain Management, Rehabilitation, & Neurosurgery teams
Lifelong Neurosurgical Concerns

1. Shunt Malfunction
2. Tethered Cord
3. Chiari II malformation

Shunt Malfunction

- Shunt Blockage
- Shunt Breakage
- Shunt Infection

Tethered Cord

- Diagnosis is made by clinical evaluation
- MRI often not helpful in patient with myelomeningocele
- Symptoms:
  - Leg/back pain
  - Regression of gross motor skills
  - Regression of bowel or bladder function
  - Frequent UTIs

Chiari II Malformation

- Downward herniation of the hindbrain into the spinal canal
- Clinical symptoms of Chiari II
  - Stridor or apnea, may require tracheostomy
  - Swallowing difficulties, may require g-tube
  - Vocal cord paralysis
  - Headaches
  - Progressive scoliosis
  - Balance & coordination difficulties
  - Tongue fasciculations (VIDEO)
- Symptom presentation earlier in life is associated with higher morbidity & mortality

Case 3: PT

- Myelomeningocele diagnosed prenatally, delivered at 38 weeks gestation via elective Caesarean
- Active Issues:
  - Hydrocephalus
  - Complex HP shunt system, s/p multiple shunt revisions
  - Chiari II malformation
  - s/p multiple posterior fossa decompression surgeries
  - Tethered cord
  - Central hyperventilation
  - Tracheostomy & ventilator dependence
  - Neurogenic bladder
  - Bladder regimen with Mitrofanoff valve
  - Neurogenic bowel
  - Bowel regimen via Chait cecostomy tube
  - Failure to Thrive
  - Nutrition via G-tube
  - Neuromuscular scoliosis
  - s/p spinal fusion

PT: Psychosocial Life

- Very strong family support team
- Lives with parents & 2 brothers
- Mother is PT’s primary caregiver
- Receives home nursing
- Graduated high school on time
- Transitioned to Life Skills Program
PT: Year 18

- Presents to ED with recent hx:
  - Lower O2 sat at baseline & frequent desaturations
  - Not tolerating any sprints off ventilator
  - Gasping
  - Worsening nystagmus
  - Worsening control of secretions
  - Tongue deviation & increased fasciculations
- What is your differential?

PT: Adult Life

- Hospitalized for work up & discussions over goals of care
- Discussions of palliative care were initiated
- Care conference to discuss quality of life
- Discussions of transition to adult care on hold
- Work up revealed severe under-ventilation
  - Tracheostomy changed
  - New ventilator
  - Discharged home with new care plan & modified DNR status

Psychosocial Concerns

- Neuropsychological development
- Vulnerability to physical, psychological, sexual abuse
- Mobility & means of locomotion
- Plans for education & employment
- Living arrangements
- Mental health
  - Depression
  - Chronic pain
- Maintaining independence

Transition Challenges

- Finding providers who care for adults with complex needs
- Care Coordination between specialists
- Myelomeningocele patients still need routine screenings
- Provider comfort with sexual counseling for patients with disabilities
- Lack of routine care can result in
  - Amputation from poor skin care
  - Nephrectomy from poor urologic management
  - Mortality from shunt malfunction

Transition to Adult Care

- AAP recommends the “transition” process starts by age 14
- Adolescents should participate in their individualized care & begin to take autonomy for their healthcare
- Parents may have trouble letting go
- Need for a “transitional program”
  - Orient young adult & family to the world of adult medicine
  - Assist with transferring medical records
  - Coordinate care between adult specialists
Summary

• Myelomeningocele population is a challenging one
• Neurosurgical interventions are often necessary throughout the life span
• Priorities for myelomeningocele patients shift as they age
• A strong multidisciplinary team is essential for this population
• Improving the transition to adult care needs to be a priority for the healthcare system

Questions?

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References