A Case of a Young Adult with Myelomeningocele: A Transition to Adult Health Care

Laura Dingfield, MD (presenter), and Allen Friedland, MD, FACP, FAAP (discussant, in italic)

Dr. Dingfield is a PGY-4 in the combined Internal Medicine and Pediatrics Residency Program at the University of Pennsylvania/Children’s Hospital of Philadelphia, and Dr. Friedland is an associate professor of internal medicine and pediatrics at Jefferson Medical College and program director of the Internal Medicine-Pediatrics Program at Christiana Care Health System.

A 21-year-old woman presents to an internist to establish care. Her past medical history is significant for myelomeningocele (spina bifida), T-10 paraplegia, neurogenic bladder status-post many surgical bladder revisions and Mitrofanoff, ventriculoperitoneal shunt for hydrocephalus, seizure disorder, migraine headaches, recurrent urinary tract infections, cecostomy for chronic constipation, and depression. A condensed summary of her medical and surgical history was prepared by her pediatric team.

Spina bifida is a congenital chronic condition that has effects on multiple systems (e.g. neurologic, orthopedic, urologic, psychologic, social, growth, genital, gastrointestinal, respiratory, and oro-motor), the family, and the health system. The defects arise by the end of the fourth week after conception and result from failed closure of the neural tube, with improper formation of the spinal cord, dorsal vertebral bodies, and overlying tissues. The long-term prognosis is based on several factors, including the location of the spinal lesion, urologic abnormalities and renal disease, and the presence or absence of hydrocephalus. As is the case with many congenital or childhood-acquired conditions, interaction between the patient and the family—and successful transition to the adult health care system—are critical to long-term prognosis.

The patient was previously cared for by a hospital-based pediatrician and pediatric specialists. Previous attempts to transition her to an adult primary care physician and urologist closer to her home had recently failed, as the physicians refused to care for her because she was “complicated” and suggested the family move to another location in order to receive specialized care.

More than a half million children with special health care needs become adults each year, and more than 90% of these individuals survive past age 30, including those with spina bifida. It is critical for providers caring for adults to have a better working knowledge of how to address the medical and psychosocial needs of these patients.

Transition of patients into the adult health care system is a challenge to patients, families, pediatricians, and providers of adult care. It is now recommended that between ages 12 and 14 certain skills be taught and reinforced with the expectation that the transition to adult care will begin between ages 18 and 21. Transition is a high-risk endeavor for many individuals with childhood acquired conditions, as it has been shown that outcomes are worse and mortality is higher in many conditions during this time period. To help facilitate safe transition of patients with complex childhood-acquired conditions, it is recommended that detailed health summaries be prepared for adult providers and that verbal signout be performed, if deemed necessary.

The patient was recently hospitalized in a children’s hospital for a urinary tract infection the week before her 21st birthday, when she was expected to transition to adult care. She was treated with intravenous antibiotics in the hospital and discharged on an oral regimen. On review of her records, she has spent 216 days of the previous two years in the hospital secondary to upper urinary tract infections. She is supposed to self-catheterize her Mitrofanoff at least every four hours while awake and irrigate her bladder with acetic acid three times per day. However, she reports that this regimen has been difficult to follow.

Individuals with congenital or childhood-acquired conditions may have comorbidities or prior treatments/procedures that are unfamiliar to providers of adult care. Knowledge of these comorbidities and standard treatments is essential. For example, neurogenic bladder with physiologic and/or anatomic abnormalities of the urinary tract is a hallmark of patients with spina bifida and is the leading cause of mortality in this population. Prevention of urinary tract infections and elevated urinary tract pressures is needed to prevent or reduce renal injury. Improvements in surgeries, clean intermittent catheterization (CIC), and medications to lower bladder pressures, minimize urinary reflux, and prevent urinary tract infections have led to increased survival.

Bladder management is required by the majority of adult patients. Most adult patients use CIC via the urethra or an abdominal stoma that enters the top of the bladder more commonly than indwelling urinary catheters. In our patient who is wheelchair bound, her abdominal stoma (Mitrofanoff or appendicovesicostomy) is the conduit from her abdominal wall to upper bladder. This allows for more privacy, easier access, and less parental dependence to comply with CIC.

The patient admits that a bout of depression preceded her most recent hospitalization, that she is now hopeful that she will have a better understanding of her care and be better able to care for herself and move forward with her adult life.

The patient wishes to move to another location in order to be closer to her family and have access to a more supportive health care system. The patient has been referred to a hospital-based pediatrician and urologist closer to her home, who recently performed a spell bind and bladder evaluation. The patient is looking forward to being able to self-catheterize, whereby she has reported to her pediatrician that it does not seem ‘as bad’ as in the past. She continues to feel hopeful and forward looking.

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She agrees to a two-month follow-up visit.

In addition to age-appropriate preventive care and surveillance, individuals with childhood-onset conditions may require additional testing or screening. Patients with spina bifida require the adult provider to consider these issues:

- Bone Health: Osteoporosis is prevalent in patients with spina bifida due to immobility. Adolescents and young adults with spina bifida may have more fractures than older adults and may present with redness and/or swelling without pain. Bone density screening is recommended, but consensus on timing of screening is lacking.

- Cardiology: The prevalence of metabolic syndrome in individuals with spina bifida is at least 30%. These patients are at increased risk of diabetes and cardiac disease.

- Functional Status: Up to 50% of ambulatory patients with spina bifida become non-ambulatory in adulthood due to altered biomechanics. Worsening kyphoscoliosis and pulmonary reserve occur as obesity and inactivity increase. Efforts should be made to encourage mobility through exercise programs.

- GI: Many suffer from neurogenic bowel and must cope with fecal incontinence, constipation, or both. Regular attention should be paid to a patient’s bowel regimen in order to ensure a predictable and regular stooling pattern.

- Nephrology/Urology: Yearly renal ultrasounds, urodynamics, laboratories, and blood pressure are recommended because a change in function may indicate a worsening neurological or urologic issue. Those patients with bladder augmentation may be at increased risk of bladder cancer in the future, but the current recommendation is against routine screening with cystoscopy and cytology for asymptomatic individuals.

- Neurology: Though most VP shunt malfunctions occur in the first three decades of life, malfunctions in adults may present with slow changes in cognition and chronic headache. New ataxia, upper extremity pain, weakness, and nerve palsies may suggest symptomatic Chiari malformation, syringomyelia, or tethered cord. Gait, orthopedic, and bowel or bladder changes may also indicate symptomatic tethering of the cord (even if this was repaired in the past).

- Ophthalmology: Many visual disturbances including strabismus, esotropia, exotropia, and orthoptopia can occur in this population. Annual ophthalmologic examination should be considered.

- Reproductive Health: Sexual education and counseling on reproductive health should be performed during the young adult years. Males with spina bifida may have impaired sexual function depending on the level of their lesion, and those with lesions above T10 are at increased risk of azoospermia. In terms of reproductive counseling, both men and women with spina bifida are at increased risk of having a child with spina bifida. Patients must be on much higher doses of folic acid to reduce this risk.

- Skin: Pressure ulcers in areas with altered sensation in the lower extremities and sacral area are common. It is important to examine wheelchair-bound patients outside of the wheelchair in order to visualize the skin well.
Since transitioning to adult care, the patient has made significant strides over the past three years. In addition to her medical issues, care has focused on providing psychosocial support and addressing the patient’s issues of social isolation, low self-esteem, and depression. She now travels (two hours each way) by para-transit to her appointments, has held several jobs, speaks about the importance of successful transition to adult care for children with special health care needs, and has not been hospitalized in more than 16 months.

**Take Home Points**

1. There is a large population of individuals with congenital and child-onset conditions that are living well into adulthood and need to transition successfully to the adult health care system.

2. Patients with spina bifida and other chronic diseases of childhood frequently have comorbid anxiety, depression, and impaired executive functioning, which negatively impacts physical health.

3. In addition to age-appropriate screening and health maintenance, patients with chronic childhood-onset conditions often require additional disease-specific considerations.

**Suggested Readings and Resources**


**Websites**

