Transition to Adult Care for Patients with Spina Bifida

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INTRODUCTION

Spina bifida (SB), Latin for “split spine,” is a congenital disorder that results from the incomplete closure of the neural tube caudally. Individuals affected with SB may have multisystem involvement, leading to complex physical and psychosocial conditions.

EPIDEMIOLOGY

Closure of the neural tube is typically complete by day 28 of embryologic age; at this time, some women may not be aware of their pregnancy. Incomplete closure of the neural tube caudally results in SB. SB occurs worldwide and across all ethnic backgrounds, although certain geographic and ethnic groups may be predisposed. 1 There is an increased risk in certain populations, including Irish and other northern Europeans; although the genetic link is unclear, it seems to be related to altered folate metabolism. 2 In the United States, approximately 1500 babies are born with SB

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KEYWORDS

• Spina bifida • Myelomeningocele • Adult • Transition

KEY POINTS

• Individuals with spina bifida have multisystem involvement, leading to complex physically impairing conditions.
• Individuals with spina bifida are living into adulthood.
• There is a tremendous need for adult-centered care for those with spina bifida.
• There are successful outcomes in transitional adult programs for individuals with spina bifida.
each year. In the United States, Latina women have the highest prevalence of having a child born with SB in comparison with other ethnic groups. The cause is unclear as to why Latina women have the highest prevalence, but it has been hypothesized that differences in dietary habits, supplement use, or social structure may play a role.

The etiology of SB is multifactorial; however, some factors that may increase a woman’s risk of having a child with SB include:

- Family history of previous neural tube defect
- Folic acid deficiency
- Altered folate metabolism
- Exposures to certain medications (including valproic acid, carbamazepine, methotrexate and other folic acid antagonists, excess vitamin A, retinoic acid)
- Alcohol consumption
- Obesity
- Fever
- Maternal diabetes mellitus

Folic acid supplementation has been shown to lower the risk of having a child with SB. In 1992, the United States Public Health Service recommended consumption of 0.4 mg of folic acid daily to women of child-bearing age to decrease the risk of neural tube defects. In 1998, the US Food and Drug Administration mandated that folic acid be added to enriched grain products, such as cereals. Since the fortification of folic acid into grain products, the United States has had a 31% decrease in the prevalence of SB, from 5.04 babies affected per 10,000 births to 3.49 per 10,000 births.

INTEREST IN ADULTS WITH SPINA BIFIDA

Medical and surgical advancements, such as antibiotics, ventriculoperitoneal shunts, and intermittent catheterization for the management of back closure, hydrocephalus, and neurogenic bladder respectively, have increased the survival into adulthood for patients with SB. From 75% to more than 85% of children born with SB survive into adulthood. With the increased longevity of children with SB new challenges arise, one of which is transitioning to adult-centered care. US Surgeon General C. Everett Koop, MD, was a strong voice for the rights of children with disabilities. In 1984, he cohosted a national conference focusing on the need for transitional care for older adolescents living with chronic childhood conditions. This conference brought the issue of health care transition onto the national radar.

Organizations such as the American Academy of Pediatrics (AAP) have published guidelines on transition of adolescent and adult care, and provision of a medical home for those affected with chronic childhood conditions, including SB. This challenge has been recognized not only in North America but around the world, including Europe and Australia. The First (2009) and Second (2012) World Congresses on Spina Bifida Research and Care brought together leaders in SB from more than 30 nations to share their experiences in different specialties and countries, and promote the development of new collaborative research ideas.

The Rehabilitation Act of 1973, the Individuals with Disability Education Act in 1975, and the Americans with Disability Act in 1990 improved integration and access for those with disabilities. In addition to the enhanced integration and access for those with disabilities, role models with disabilities and mentors have played a role in providing the sense of possibility and success. Such an exemplar is Tatyana McFadden, 11-time medal winner in Summer and Winter Paralympics and the person...
behind a landmark legislation requiring public schools to provide access to sports for students with disabilities in Maryland. This law is now a model for other states.

**TRANSITIONAL CARE**

Multidisciplinary clinics are the standard of care for children with SB in the United States. To maximize medical and functional outcomes it is essential for physiatrists, neurosurgeons, urologists, orthopedic surgeons, and primary care physicians to communicate and collaborate.21

As those with SB age, unique challenges arise. The transition to adult health care has lagged behind the medical advancements that have increased survival into adulthood. Discussions on transitioning care should occur during times of relative wellness, and not during an acute medical crisis.22 One of the greatest barriers to transitioning to adult health care was finding access to health care providers who took care of adults with special needs, in both the primary care setting and medical subspecialties.23,24 Transition should be a gradual process and be a coordinated approach, with the adolescent/young adult taking responsibility for the direction of his or her care with familial support.25 The recommended age for starting the discussion on transition is 14 years, as recommended by the AAP. Fourteen is also the age at which one’s Individualized Education Plan for postsecondary transition planning begins. However, if the child is developmentally appropriate and can understand his or her medical condition, and can demonstrate adherence to treatment plans, the discussion on transition can begin at an earlier age.

A transition study conducted in Canada identified youths and adults with SB, cerebral palsy (CP), and acquired brain injury (ABI). The experiences and opportunities of these unique individuals and groups were studied. Although the sample size was small for the group of adults with SB (13), the study did highlight that the adults with SB, when compared with those with CP and ABI, was the most highly educated group, had the highest percentages living alone (23.1%), and had the highest percentages of those working full-time (30.8%). However, those adults with SB had the worst self-rated health of all the subgroups.26

In a 25-year prospective study on the outcome of those with SB in a multidisciplinary clinic, medical needs and successful outcomes were chronicled. Patients underwent shunt revisions, scoliosis surgeries, and tethered cord releases. Of those who underwent tethered cord releases, 97% had improvement or stabilization of their preoperative status. More than 80% of the young adults with SB had social continence of the bladder, with 85% of them having attended or graduated from high school or college.14 On the other hand, a retrospective telephone study was done on a cohort of patients with SB, who were previously seen in a multidisciplinary clinic that disbanded. Three years after the clinic disbanded, no one had coordinated their care, and up to 66% of patients with SB did not have regular medical and specialty care. There was an increase in serious morbidity, including amputation and nephrectomy.27 Transitioning care is critically important for maintaining the health of adults with SB.

**INDIVIDUALS WITH SPINA BIFIDA HAVE MULTISYSTEM CONDITIONS**

**Primary Care**

Finding a primary care provider that is knowledgable and interested in taking care of adults with special needs is a challenge. Collaboration has been made between the AAP, American Academy of Family Physicians, American College of Physicians, and the American Society of Internal Medicine in the “Consensus Statement on Health Care Transitions for Young Adult with Special Health Care Needs.”16
Young adults have a higher prevalence of being classified as overweight or obese. Adults with SB, as with able-bodied adults, require regular primary care visits with an adult-oriented physician for annual physical evaluations, routine preventive cancer screening examinations (Pap smears, breast examinations, testicular examinations), and monitoring for metabolic syndrome. Individuals with disabilities have been noted to have worse outcomes with cancers because of late detection and treatment.

**Neurosurgical Care**

Neurosurgeons are some of the first physicians an infant with SB and family will meet after delivery/birth. In addition to the neural tube defect, the infant may have hydrocephalus or a Chiari malformation. Hydrocephalus in infancy is significantly different from hydrocephalus, which begins in adulthood. In addition to the adult causes of hydrocephalus (tumor, meningitis, hemorrhage), infants can develop hydrocephalus from abnormal brain development, which obstructs the flow of cerebrospinal fluid (CSF), and there may be multiple points of potential obstruction of the CSF. Shunt failure can happen at any time, including adulthood. Up to 25% of patients with infantile hydrocephalus can have shunt failure in adulthood with nonresponding ventricles (i.e., no change in the size of the patient’s ventricles, but the patient has markedly increased intracranial pressure). Patients with SB and hydrocephalus have increased mortality as adults, mostly due to shunt failure. As patients and families transitioned, often they were most concerned about shunt malfunction.

In addition to shunt malfunctions and Chiari complications, tethered cords and syringomyelia are other potential medical complications of adolescents and young adults with SB. Tethered cord can be a delayed complication of those with SB, usually occurring during late childhood or adolescence. However, the need for untethering in adults has been documented in the literature.

When a poll was taken at the 2008 meeting of the American Society of Pediatric Neurosurgeons, 52 practitioners (approximately half of the membership) reported working at a facility in which they were unable to care for patients beyond a certain age. Many of them worried about the effect the transition would have on their patients, and 36% of the pediatric neurosurgeons did not have an identified general adult neurosurgeon to whom to transfer their patients. During the transition to adult care, some patients may not have an established relationship with an adult provider in neurosurgery. Therefore, physiatrists may play a pivotal role in the referral process and in providing key information to a neurosurgeon about changes in function. This coordination can assist the surgeon in deciding if or when to proceed with surgical intervention in a given patient, as shunt or tethered cord symptoms may present with less obvious findings.

**Urologic Care**

Urologic care is focused on the preservation of renal function and promoting urinary continence (social continence). Neurogenic bladder complications that can occur with patients with SB include elevated detrusor leak point pressure, vesicoureteral reflux, and detrusor-external sphincter dyssynergia. Current bladder management for children with neurogenic bladder in SB emphasizes lowering the urinary storage pressure with anticholinergic medications and using clean intermittent catheterization to avoid elevated voiding pressures. With compliance in taking anticholinergic medications and performing clean intermittent catheterization, up to 92% of children demonstrated normal renal function. Botulinum toxin injections have also been used on the bladder to increase capacity.
As the patient transitions to adolescence, a small study found that puberty was associated with an increase in maximum cystometric capacity, detrusor pressure, and detrusor leak point pressure, owing to prostatic growth in males or estrogenization of female urethras.35

Sexual counseling should be included in the adolescent/young adult clinic visit. In a survey of adolescent/young adult patients with SB, only 52% were satisfied with their sexual lives. Women with SB were 2.3 times more likely to be sexually active than men. There is a high prevalence of erectile dysfunction in men with SB.36 When discussing sexuality with the patient, bladder and bowel concerns should be addressed, such as bladder leakage or bowel accidents during intimacy. Discussion should also include the issue of latex allergy and (latex-free) condom use.

In one study, up to two-thirds of adults with SB did not have regular urologic follow-up.37

In adults with neurogenic bladder in SB, there are no established guidelines for urologic surveillance. However, in the absence of urologic disorder, a baseline video urodynamics study and repeat every 2 to 3 years, annual renal ultrasonography, and annual serum creatinine level is recommended. Any change in urologic care and/or function warrants a repeat urodynamic study; consider tethered cord in the differential as well, if there is a change in bladder care/function. If there are complications, such as recurrent urinary tract infections (UTIs), pyelonephritis, stones, hematuria, abnormality on ultrasonography, unresolved vesicoureteral reflex, or progressive renal compromise, consider further evaluation with voiding cystourethrography and nuclear medicine renal function scans.35

In a case series, 8 patients with SB, neurogenic bladders, and chronic UTIs developed bladder malignancies.38 There may also be an increased cancer risk with augmentation cystoplasty, with a 1.2% incidence of bladder cancer in augmented bladders in children. For these patients, an annual cystoscopy and cytologic examination of urine is recommended.39

In those with chronic UTIs, most interdisciplinary clinics treat with antibiotics when there are greater than 10,000 CFU/mL in association with:

1. Urine white blood cell count greater than 50 WBC/microliter of urine
2. Fever
3. Flank pain
4. Dysuria, change in urinary pattern40

Those who undergo bladder reconstructive surgery as children continue to have complication risks into adulthood. Risks may include stomal complications, stones, ureteric stenosis, or bladder rupture.

Orthopedic Care

Many children with SB have orthopedic complications, including club feet, dislocated hips, scoliosis, and contractures. Some of these orthopedic conditions may progress over time. Foot deformities are the most common orthopedic abnormality in children with SB. As the patient ages, calcaneovalgus deformity is commonly seen.41 If there is a change in the foot position and neurologic status, consider tethered cord or syringomyelia. Individuals with SB are at increased risk for low bone mineral density, hence increasing their risk for fractures.42 Overuse injuries of the shoulder were found to be more prevalent in those propelling wheelchairs than in those using crutches. Medial knee pain from valgus stress resulting from long-term abnormalities in gait may be ameliorated with forearm crutch use (First World Congress on Spina Bifida Research and Care).
Physiatrists are uniquely trained in taking care of individuals with complex, physically impairing conditions. A person’s functionality is greatly affected by the level of involvement of their SB. Motor level and a history of hydrocephalus significantly affect a patient’s mobility. Young adult-onset shunt failure, tethered cord, syringomyelia, neuro-orthopedic problems, obesity, and premature skeletal aging can all contribute to functional declines and loss of ambulation in this population.\(^\text{33}\) Despite having high-quality assistive devices, adults with SB who use wheelchairs had a lower activity level, both in the home and outside the home, in comparison with those who ambulate.\(^\text{43}\)

Physiatrists can assist in the transition process by promoting and maximizing the young adult’s ability to self-care, assisting with integration to school and work, and supporting activities of interest.\(^\text{44}\) Responsibilities for self-care may include management of bladder and bowel programs, ordering supplies, making medical appointments, arranging transportation, and participating in the meetings for one’s individualized education plan. Aside from actively encouraging self-management, the physiatrist can also assist the patient in promoting self-advocacy.

**Gastrointestinal Care**

In individuals having neurogenic bowels, the areas mostly affected in children with SB are the large intestine, rectum, and internal and external sphincters. Encouraging social continence with a bowel program is recommended. Eighty percent of those with SB need to be utilizing some form of bowel program. Implementing routine/regular schedule, increasing liquid intake, timed bowels, disimpaction, digital stimulation, the gastrocolic reflex (it is unknown if this reflex is intact in those with SB), retrograde enemas, and taking stool softeners or bulking agents may help.\(^\text{41}\) Surgical anterograde continence enema may be an option for those with intractable incontinence.

**Cognitive and Psychological Care**

Children with SB have good verbal skills, but may have learning disabilities and difficulties with problem-solving tasks, such as decision making about treatment plans,\(^\text{45}\) which can affect self-management of care. A retrospective chart review documented that there was a delay of 2 to 5 years in self-care or autonomy of adolescents with SB.\(^\text{46}\) One study recommended neuropsychological evaluations during key developmental transitions (during early childhood, early adolescence, and adolescence/young adulthood)\(^\text{47}\); however, obtaining authorizations for these evaluations may prove difficult.

As children with SB become older, self-awareness occurs. Children may become more concerned with body image. It has been found that adolescents with SB who have low body image had a propensity for depression. Those who were not socially continent had lower self-esteem.

Gender and disability may place women with disabilities at greater risk for psychosocial problems, such as depression.\(^\text{48}\) These women are also vulnerable to physical and psychological abuse, in addition to sexual exploitation.\(^\text{49}\)

Full-time wheelchair users had lower scores on their quality-of-life survey, yet psychological distress symptoms did not vary in comparison with other groups.\(^\text{43}\)

For many parents with special needs children, caring for their child has been a full-time job. Some parents may feel uneasy about “letting go” as their child transitions from the pediatric health care clinic to the adult health care system.
Resources for psychological support for both patient and parents should be provided.

**Women’s Health**

As with the general transition of care from adolescents/young adult to adult-centered care, it is a challenge to transition a young woman’s health care to adult providers who are knowledgeable and interested in seeing a woman with special needs. Access to Pap smears and mammograms is difficult. The importance of folic acid supplementation should be emphasized with all women of child-bearing age and their first-degree relatives (parent and siblings), and female partners of men with SB. All women of child-bearing age should take supplements of folic acid of 0.4 mg/d. Those with previous pregnancies resulting in neural tube defects should take 4 mg/d.

Young women with SB who are pregnant should be followed by a high-risk obstetrician. Pregnancy may affect bowel and bladder care as the gravid uterus progresses. Depending on the level of involvement, it is possible that the patient may not be able to feel contractions; therefore, education on other signs of labor should be discussed (eg, rupture of membranes may cause leakage).

**Social, Living, and Vocational Concerns**

As patients with SB become adults, the need arises for independence in life skills, self-management, housing, and employment opportunities. Executive function issues can interfere with initiation and problem solving. The Department of Vocational Rehabilitation is available at the county or state level. Individuals can be referred to local regional centers, which may have social workers or resources available for those with disabilities. The local chapter of the Spina Bifida Association is also an asset. Local support groups or sports-related organizations for challenged athletes can help with social transition.

**TRANSITIONAL PROGRAM**

Developing a transitional program takes a coordinated effort involving multiple departments, health care providers, and dedicated individuals. One such transitional program had the patient meet with a nurse to orient him or her regarding the transition, summarize the medical history, had the patient tour the adult care facility, transfer medical records, and undergo transition visits at the adult clinics. The health care providers at the adult facility were involved in the adult spinal cord injury program and consisted of the departments of Physical Medicine and Rehabilitation and Urology. Although there were initial reservations about the transition, 82% of the participants had a “positive experience” even though it was difficult to engage and establish rapport with a new team of providers. Patients generally appreciated being “treated like an adult.” Patients and families wanted more resources on psychological services, employment and housing opportunities, obtaining adaptive equipment, and resources on nutrition and physical activities. Some concerns of patients and families included the lack of routine neurosurgical and orthopedic follow-up, and lack of effective communication. A focus group for young adults with SB in Chicago showed similar results, with individuals expressing appreciation that they were “treated like an adult,” were given decision-making control by the treatment team, and looked forward to the challenge of increased responsibility for their care. However, some parents expressed concerns about losing lifelong relationships with the pediatric providers and not having as much control over medical decision making.
Five key elements for a transition program were highlighted:

1. Preparation
2. Flexible timing
3. Coordination of care
4. Transition clinic visits
5. Health care providers interested in taking care of adults with disabilities

**SUMMARY**

Individuals with SB are living into adulthood, and as they age unique challenges arise. These patients have involvement of multiple organ systems, physical impairments and disabilities, cognitive involvement, and psychosocial challenges. There is a tremendous need for transitional care for adults with SB.

**RESOURCES**

- Centers for Disease Control and Prevention: [www.cdc.gov](http://www.cdc.gov)
- Spina Bifida Association: [www.spinabifidaassociation.org](http://www.spinabifidaassociation.org)
- SB University: [www.SBUUniversity.org](http://www.SBUUniversity.org)
- World Congress on Spina Bifida: [www.worldcongressonsb.org](http://www.worldcongressonsb.org)
- American Latex Allergy Association: [latexallergyresources.org](http://latexallergyresources.org)

**REFERENCES**


