Chapter 33
Spinal Dysraphism: Impact of Technique and Technology on Expectations

David G. McLone, M.D., Ph.D.

The history of spina bifida stretches back to ancient times; however, only recently has the plight of an infant born with an open neural tube defect improved significantly. Before the middle of the last century, few infants survived, and those that did led difficult lives.

In the 1950s, a son was born with spina bifida to the Holter family. John, the father, an engineer, developed the first effective differential pressure valve to control hydrocephalus (11). As often happens, this was perceived as the ultimate answer to the problems of the child with spina bifida.

In the 1960s, surgeons around the world treated the birth of an infant with spina bifida as an emergency. Open neural tissue was surgically covered regardless of the time of day or night. Ventricular atrial shunts were installed to control hydrocephalus. The early mortality rate decreased rapidly, almost reversing the previous 90% mortality to a survival rate of 80% (Fig. 33.1).

As the initial exuberance wore off in the 1970s, it became clear that the shunt was an answer that exposed a number of problems not anticipated. Incontinence, paraplegia, and scoliosis coupled with the new problems of shunt malfunction, infection, and mental retardation overwhelmed the medical team. The magnitude of these problems and their impact on the children and their community caused a rethinking of our approach to this birth defect.

John Lorber (6), a neurologist in England, led the drive to control the problem. Selection criteria were developed based on the perceived quality of life expected for the infant. Children selected for nontreatment were not allowed to survive. The mortality rate climbed to 70%, and this method of caring for the newborn with spina bifida spread around the world.

A prospective study performed between 1975 and 1979 on the outcome of 100 children with spina bifida treated aggressively from birth clearly demonstrated that the selection criteria were not valid predictors of the functional outcome for the child (8). Lorber's selected population was not more functional than the unselected aggressively treated group. The only difference was the number of survivors in the selected group was much fewer.

One of the most significant contributions to the quality of life and survival for these children comes from our pediatric urological colleagues. Many children survived infancy because of the shunt, only to die from renal failure. Clean intermittent catheterization (CIC) almost eliminated renal loss, obviated urinary diversion to the abdominal wall, and allowed these children to be dry, and this led to more social acceptance and mainstreaming in our educational system (2).

The peak incidence for spina bifida births in the United States occurred in 1948. Since 1948, there has been a gradual decrease in the incidence of approximately 5% per year. The reasons for this are unclear but thought to be the result of improved nutrition for our population. The discovery of alpha fetal protein (AFP), first in amniotic fluid and

later in maternal blood during pregnancy, enabled the prenatal screening for a fetus with a neural tube defect (10). Ultrasound and amniocentesis allowed conformation, and this led to an ethical debate surrounding the abortion issue. Today, it is estimated that about 22% of affected fetuses undergo termination. For those families that elect not to terminate the pregnancy, and this information allows an informed anticipation of the birth of a child with spina bifida.

The story of folic acid use is amazing. Hibbard and Smithells (3) suspected that women who had children with neural tube defects were deficient in folate. Women given folic acid had a much lower incidence of recurrence. Today, we know, thanks to randomized prospective studies, that folic acid supplementation can reduce an expected incidence of spina bifida by 70%. In our splotch mouse laboratory model, folate supplements prevented the neural tube defect. This is indeed remarkable. A one-a-day vitamin can block the phenotypic expression of a gene known to cause a birth defect. The problem that remains is convincing all women of reproductive age to take this vitamin.

Orthopedic surgeons, like Luciano Dias (1), led the way in developing procedures to correct hip, knee, and foot deformities in these children. Many of the procedures developed for cerebral palsy were not appropriate for children with spina bifida. Most children are now upright and ambulate during early childhood. Half of young adults with spina bifida will elect to use the wheelchair to move about in the community. Spine surgery stabilized sagittal balance in children, enabling proper setting and ambulation and preserving respiratory capacity.

The multidisciplinary team approach developed by Shurtleff (13) and others in the field had a profound effect on care and outcome. Bruce Kaufman (4) would later compare populations observed in a multidisciplinary clinic with a group with fragmented care and found that deterioration was many times more likely in the group not observed by a team. In fact, the adolescent with spina bifida was eight times more likely to have a deteriorating condition.

From a neurosurgical prospective, obviously the shunt has been the major advancement in caring for individuals with spina bifida. Whereas the shunt has enabled meaningful survival for the vast majority, it continues to inflict injury on some (Fig. 33.2). Shunt infections remain a problem that cause intellectual developmental delay, and shunt malfunction all too often leads to neurological defects or death.

Reigel's technique (12) for repair of the open myelomeningocele offers an anatomically correct repair with the hope that it may also reduce the possibility of retethering of the spinal cord (Fig. 33.3). Even if the repair does not prevent or decrease the incidence of retethering, the untethering procedure is made much easier after this repair.

The progress made in the development of neuroimaging is remarkable and has had an impact on all of neurosurgery. Pneumoencephalograms, ventriculograms, nuclear scans, angiography, computed tomography scans, ultrasounds, and magnetic resonance imaging (MRI) have benefited individuals with spina bifida and enabled the neurosurgeon to provide improved care. Technical progress in instrumentation such as the shunt, magnification, bipolar, and the laser has also made a difference. (Table 33.1)

Clinically, time and observation have taught us a great deal about managing the problems of individuals with spina bifida face. Initially, many thought that if a child survived beyond the age of 4 years, most major hurdles would be behind them. This obviously is not true. Late deterioration from a variety of problems will continue to threaten the ability the adolescent and young adult to participate in our society. We now know that deterioration is almost always from a treatable cause, and the first suspect should always be the shunt. The next most common cause of

deterioration is tethered cord (12). Hindbrain, cranial nerve, and cerebellar signs (Chiari II symptoms) usually respond to either shunt insertion or revision posterior cervical decompression, often incorrectly referred to as posterior fossa decompression.

Routine MRIs have demonstrated that hydromyelia is acquired because of untreated hydrocephalus or at the time of the first shunt malfunction and usually resolves after the insertion or revision of the shunt (5) (Fig. 33.4). Longstanding holocord hydromyelia may persist in the face of a functioning shunt in an individual who is neurologically stable. In this situation, direct shunting of the hydromyelia cavity seems to offer little.

From the laboratory studies has come a better understanding of the mechanisms of neural tube closure and the effect of the open neural tube on the developing embryo. Initially, light and electron microscopy revealed the structure and interaction of the mesencyme with the neural epithelium that lead to closure. Biochemical studies showed that these processes are energy dependent. It is now known that the persistent opening in the neural tube causes the Chiari II malformation (9) (Figs. 33.5 and 33.6). This study predicted that early fetal repair of the open neural tube would improve or prevent the Chiari II malformation.

The amazing rapid progress in molecular genetics has opened a whole new area for study. Many of the genes and their interaction in assembling the embryonic nervous system are now known. As these discoveries move into the clinical scene, we can expect improved counseling of prospective parents, more accurate diagnoses, more folate-like compounds to prevent neural tube defects, and maybe even someday molecular or stem cell repair will rescue the embryo or fetus.

TABLE 33.1. Summary of progress over time[a]

	1960	1970	1980	1990	2000	2005
Mortality	90%	70%	25%	10%		
Advances	Team	AFP	Folate			
Technology	VA shunt	VP shunt	Endoscopes	Ventriculostomy		
Imaging	PEG		Angiograms	CT scans	Ultrasound	MRI
Technique	Spine CIC	Reigel	Closure	Teth cord	Intrauterine	
Laboratory	Structure	Cause of Chiari II	Molecular genetics			

[a]AFP, alpha fetal protein; VA, ventriculoatrial; VP, ventriculoperitoneal; PEG, pneumoencephalograms; CT, computed tomography; MRI, magnetic resonance imaging; CIC, clean intermittent catheterization; Teth, tethered.

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FIG. 33.1 Children born in the 1970s with a myelomeningocele and hydrocephalus.

FIG. 33.2 Some of the problems with shunts for hydrocephalus.

FIG. 33.3 *A–G*, the sequence of repair of the myelomeningocele.

FIG. 33.4 MRI showing preoperative holocord hydromyelia and postoperative resolution after shunt revision.

FIG. 33.5 *A*, normal embryo with closed neural tube (*arrow*) and expanded cranial vesicles, (*T*). *B*, open neural tube and nonexpansion of the vesicles (*T*).

FIG. 33.6 *A*, normal embryo with open posterior fossa vesicles and brain contents above the foramen magnum. *B*, embryo with an open neural tube, collapse of the posterior fossa vesicles and herniation of the lower brain stem out of the posterior fossa.