



A study of the clinical profile and outcome of spina bifida

Theophilus Nikita Kumar and Ramesh M.K

General Surgery Department, Bangalore Medical College and Research Institute
Correspondence Email: nixie_reading@hotmail.com

ABSTRACT

Neural tube defects (NTDs) are a group of congenital anomalies characterized by defects in dorsal midline structures, including neural tissue, dura, muscle, bone and/or skin. The clinical presentations and the follow-up of these patients requires attention to various end organs besides the nervous system. To evaluate the clinical profile and surgical outcome of children with spina bifida. Out of a total of 74 patients treated at our institute for spina bifida between June 2013 to August 2015, 74 cases of spina bifida were analyzed retrospectively and prospectively. The clinical profile, radiological findings and urodynamic studies were recorded. Craniospinal MRI was done in patients to screen for Arnold Chiari malformations and monitoring of hydrocephalus was done as a management protocol at our institute for these children. All these patients except two underwent surgery for correction and closure of the spinal defect. Associated anomalies were treated accordingly. They were clinically assessed over a mean follow up period of 1.3 years, ranging from 2 months to 2½ years. 73% of the patients presented in the neonatal age group. Of which, 72% presented with a visible sac over the back. 72% of the cases were Myelomeningocele. 79% of the defects were in the lumbosacral region. 30% presented with sensorimotor loss or bladder bowel incontinence. Sensorimotor improvement was seen in 12.5% after repairing the defect with the help of physiotherapy and braces. 30% of the patients were diagnosed to have hydrocephalus, of which 33% required a CSF diversion procedure. The postoperative course of spina bifida repair was found to be uneventful in 90% of the patients.

Key words: Neural Tube Defect, Spina Bifida, Myelomeningocele

INTRODUCTION

NTDs are among the most common of all human birth defects. The incidence of spina bifida is estimated at one to two cases per 1000 population, with certain populations having a significantly higher incidence based on genetic predilection. There is also a marked geographic variation in incidence. [1]

Defects of the neural tube encompass a wide range of congenital spine and spinal cord defects. These defects involve the imperfect development of the neuropore during embryogenesis and the subsequent maldevelopment of the adjacent bone and mesenchymal structures. [2]

The primary embryological defect is believed to be a failure of, or incomplete, neural tube closure. This is a process that normally occurs during the third to fourth week of fetal life. These lesions can involve any part of the spine, although they most often involve the lumbosacral spine, and range from a simple gap in the lamina of a single vertebral level to an extensive dorsal opening with an exposed spinal cord. [3]

Neural tube defects are commonly classified as open or closed on the basis of the presence or absence of exposed neural tissue. Open neural tube defects, include anencephaly, spinalrachischisis or spina bifida aperta/cystic (i.e., myeloschisis, myelomeningocele, and meningocele), and encephalocele. Closed neural tube defects include spina bifida occulta which include, lipomatous malformations (e.g., lipomas and lipomyelomeningoceles), split cord malformations (diastematomyelia, diplomyelia), neurenteric cysts, dermal sinuses, tethered spinal cord, and sacral agenesis (caudal regression). Of the open neural tube defects, myelomeningocele is the most common and the most severe birth defect compatible with survival. [4]

The myelomeningocele and associated hydrocephalus were known to Hippocrates, Aristotle, and other ancient physicians. In the early twentieth century, surgical techniques had progressed to allow the closure of open defects without immediate perioperative mortality caused by infection, but the untreated hydrocephalus led to impaired mental and physical function in most of the survivors. With the effective treatment of hydrocephalus by shunting in the 1950s, most myelomeningocele patients received aggressive care. As they survived, however, many continued to suffer from significant physical and mental disabilities, such as deformity of the extremities, severe scoliosis, shunt infections, and significant urinary dysfunction and failure. [5]

Severe spinal dysraphic disorders constitute a major source of disability among children and adults. Management of spinal dysraphic anomalies involves a number of steps: accurate diagnosis, an assessment of the severity of the lesion, a decision whether intervention is warranted, the nature of the intervention, and educating the family of the need for lifelong medical care.

But to do so, surgeons need reliable data regarding presentation and outcome in order to help parents faced with difficult decisions about termination of an affected pregnancy or treatment after birth.

MATERIALS AND METHODS

This study was conducted over a period of two and a half years, from January 2013 to June 2015, in the Pediatric Surgery Department, PMSSY Hospital, Bangalore Medical College and Research Institute, Bangalore, India. 74 patients were included in the sample study and were analysed both retrospectively and prospectively. An informed written consent was taken from each patient's attenders. After which, a clinical examination was done to assess for sensorimotor function, orthopaedic anomalies and bladder and bowel function, besides taking into account the spina bifida defect, and the results were documented.

X-rays of the spine and neurosonograms were done to assess occult anomalies if present. Patients were subjected to MRI brain to diagnose Arnold Chiari malformations, Dandy Walker anomaly, ventricular volume, V/H ratio and mantle thickness.

Ultrasonogram KUB was done to assess for hydronephrosis, vesicoureteric reflux and contrast studies were done to rule out gastrointestinal anomalies.

72 patients underwent surgical correction of the defect post-delivery. Patients were operated within the 48 to 72 hours of initial presentation. Patients with myelomeningocele underwent a myelomeningocele repair and patients who presented with symptoms of tethered cord or a spina bifida occulta underwent a detethering and a laminectomy.

These patients were followed up over a mean follow up period of 1.3 yrs ranging from 2 months to 2 ½ yrs. They were followed up every 3 months postoperatively where they were assessed for head circumference, sensorimotor function improvement or deterioration and bladder and bowel function. Shunt function was checked in patients who had undergone CSF diversion procedures. At each follow up visit patients underwent serial neurosonograms for documentation of ventricular volume, V/H ratio and if required shunt procedures were revised or freshly placed based on clinical manifestations (mechanical or infective causes), ventricular volume.

Patients with bladder incontinence were subjected to urodynamic studies and advised either surgery or clean intermittent catheterisation based on the cause of incontinence. As the patients we dealt with primarily had overflow incontinence with negative bladder neck pathology as concluded by urodynamic studies, no surgical interventions were planned and patients were advised clean intermittent catheterisation. Bowel incontinence was dealt with bowel enemas, laxatives and diet modifications.

STATISTICS

This study was a cross sectional study. Analysis was carried out based on mean, median and mode. Rates, ratios and percentages were taken into account to express most of the data.

RESULTS

From January, 2013 to June, 2015, 74 patients were diagnosed with spina bifida.

Age of presentation: 73% of these patients presented in the neonatal age group.

Mode of presentation: 72% presented with a visible sac over the back and a minority presented with neurological deficits and dermal markers.

22 patients out of 74 (i.e. 30%) presented with neurological deficits. 22 out of 74 patients presented with neural deficits. 4 patients had only lower limb motor and sensory loss. 2 out of these 4 showed Grade 2 paraparesis where as the other 2 had complete paraplegia.

6 patients presented only with bladder bowel incontinence with no signs or symptoms suggestive of lower limb weakness.

12 out of 22 presented with a combination of paraplegia and bowel and bladder incontinence. 2 out of 21 patients of spina bifida occulta presented with a hairy patch and the other with a dermal sinus.

Type of neural tube defect seen: 72% of the patients had a myelomeningocele and 28% of them had a spina bifida occulta. 79% of the patients had a lumbosacral defect.

10 out of 21 presented with neurological deficits which on investigation concluded to be cases of spina bifida occulta. 9 patients presented with lipomyelomeningoceles.

Association with Hydrocephalus: 21 out of 74 (i.e. 30%) patients developed hydrocephalus. 15 were diagnosed with hydrocephalus before the surgical repair of the defect and 6 developed hydrocephalus postoperatively which was diagnosed during follow up. 33% of the hydrocephalus patients underwent a CSF diversion.

Postoperative course: Postoperative course of spina bifida repair was found to be uneventful in 90% of the patients. Surgical site infection was seen in 2 out of 72 patients, intraoperative nerve damage in 4 out of 72. Neither improvement nor progression of sensorimotor loss was seen in the majority of the patients postoperatively.

Follow up: Patients were followed up over a mean period of 1.3 years which ranged from 2 months to 2½ years. Sensorimotor improvement was seen in 12.5% through continuous physiotherapy and use of braces. All patients started on clean intermittent catheterisation and bowel washes could be kept dry by day and night if found to be compliant, i.e clean intermittent catheterisation by day and continuous drainage at night. Retethering was seen in 1 patient over a follow up period of 1 year and was dealt with by detethering.

Mortality: 2 out of 74 patients died preoperatively due to aspiration and sepsis.

DISCUSSION

The incidence of spina bifida is estimated at one to two cases per 1000 population, with certain populations having a significantly higher incidence based on genetic predilection. There is also a marked geographic variation in incidence. The highest rates occur in parts of the British Isles, mainly Ireland and Wales, where the incidence of myelomeningocele is as high as three to four cases per 1000 population.

There are two fundamental theories regarding the embryogenesis of myelomeningocele, both encompassing a disorder of primary neurulation. In the so-called nonclosure theory initially suggested by Von Recklinghausen, it is proposed that neural tube defects represent a primary failure of neural tube closure. In the over distension theory, introduced in 1769 by Morgagni and popularized by Gardner, it is proposed that neural tube defects arise through over distension and rupture of a previously closed neural tube. The non-closure theory is more widely accepted and certainly accounts for the majority of human neural tube defects. [4]

Genetic factors do seem to have a role in some cases. The risk of one child having spinal dysraphism is estimated at 0.1% to 0.2%, but with one affected sibling the risk of a second affected child increases to 2% to 5%, and the risk of a third affected child increases again to 10% to 15%. These occurrences do not fit a Mendelian pattern of transmission. Other genetic mechanisms of transmission, such as an X-linked recessive gene, a dominant gene with variable penetrance, or polygenic transmission, have been suggested to explain this tendency to recur within families. [5] In our study none of the parents gave history of a previously affected child and hence genetic co relation was not assessed in our study.

Whilst the incidence of meningocoele and myelomeningocoele is well documented, there is a paucity of data concerning the frequency of spina bifida occulta. This is attributable to two main factors. Firstly, the condition is

usually symptomless and therefore often passes undetected. Secondly, some radiologists regard spina bifida occulta as a normal anatomical variation and do not record it in their reports. [6]

In a study conducted in Lucknow, India, meningomyeloceles accounted for 72% of the cases, meningocele in 2%, myelocystocele in 2% and 23% had occult spina bifida [split cord malformation without overt MMC sac 19% and midline dermal sinus in 4.5%]. [7]

A lower incidence of split cord malformation (7% of the cases) and dermal sinus (1% of the cases) of was recorded by us. Meningomyeloceles accounted for majority of the cases and accounted for 72% of the cases which is similar to literature gathered.

Myelomeningocele present with a sac with neural tissue that can be demonstrated. It is usual non reducible, though a cough impulse and cross fluctuation can be demonstrated if the fontanelle is open. They may present with a moderate to severe degree of neurological deficit or even complete loss of neuronal function. The tethering effect of a myelomeningocele can lead to further deterioration as the child grows. The urinary tract may be affected in two ways, a true developmental malformation in about 20%, but more commonly neurogenic dysfunction. [8]

A number of cutaneous findings are markers for some of these underlying hidden dysraphic defects. There may be a hairy patch, a nevus, an appendage or skin tag, or a small dimple with a pinhole.

Hydrocephalus develops in approximately 60% to 85% of myelomeningocele patients, with 5% to 10% of patients having clinical evidence of hydrocephalus at birth. Hydrocephalus, if not present at birth, can also develop after myelomeningocele closure, as surgery eliminates a route for CSF egress. Definitive criteria for shunt placement vary between institutions. The overall rate of ventricular shunting is 81%. The level of the lesion significantly affects the incidence of shunting, with more cephalad lesions correlating with higher rates. [9]

As most of the patients have bladder and bowel incontinence, keeping them dry forms a vital part of well being and independent living and these issues need to be addressed individually.

Pseudo- bowel continence can be achieved in 69% incontinent patients. Of the 69%, 87.5% performing retrograde enemas, 80% performing orthograde enemas through an antegrade continence device and 8 of 10% performing regular manual evacuation of stools can be kept pseudo continent. In 20% patients performing orthograde enemas complications can lead to closure of the antegrade continence device. Success of treatment is not related to level of spinal lesion or degree of mobility. [10]

Through regular bowel enemas, laxative use and dietary modification all patients could be kept pseudo bowel continent.

Prompt detection of children with high-risk urological dysfunction and early intervention is key in protecting against hydronephrosis, preserving renal function and preventing the development of a poorly compliant bladder. In the case of sphincter over activity with detrusor underactivity, clean intermittent catheterisation is the only management needed to control urinary tract damage and leakage. Beginning CIC early in infancy conveys several advantages over expectant treatment, including easier caregiver and patient adaptation to the routine and less need for augmentation cystoplasty. In addition to CIC, pharmacological treatment in the form of the anticholinergic oxybutynin is used to treat both forms of neurogenic bladder sphincter dysfunction involving detrusor overactivity, long-term studies having demonstrated safety in children and infants and efficacy in lowering filling pressure, increasing bladder capacity, and preventing renal damage. [11]

Without treatment, historical data suggest that only 15% to 30% of myelomeningocele patients survive infancy. Current standards of care have improved the survival rate to approximately 85%, although approximately 10% will die before 6years of age, primarily due to complications from hindbrain dysfunction related to the presence of a symptomatic Chiari II malformation.

Operative mortality is practically absent, while morbidity may be significant. The most frequent complication is wound breakdown usually secondary to CSF leak, which is known to be an adverse factor for wound healing. Conversely, wound infection is a much rarer complication of MMC repair, occurring in less than 2% of procedures. The most severe, though rare complication of surgery is meningitis with sepsis, which remains the main cause of death in these newborn children. Intravenous antibiotics are the treatment of choice, and ultimately CSF shunt removal if already implanted. [12]

Intrauterine repair of myelomeningocele has been advocated as a means of improving the neurologic outcome and reducing hindbrain herniation in infants with myelomeningocele. This is based on the idea that secondary damage and resultant disability, which may occur when exposed neural tissue is in contact with amniotic fluid, may be reduced or even completely eliminated by closing the defect as early as possible in utero. However, in contrast to postnatal repair, prenatal surgery was associated with an increased risk of preterm delivery, as well as uterine dehiscence at delivery.

Treatment of NTDs has evolved over the past 50 years. Previously, neonates with NTDs either were left untreated or were treated selectively; however, most of them died of meningitis, hydrocephalus, and/or sepsis. In contrast, during the past 3 decades, neonates with NTDs have received prompt, aggressive treatment in almost all pediatric centres. Such treatment including early closure of open NTDs and aggressive shunting of hydrocephalus leads to survival with nearly normal intelligence in many patients. The multidisciplinary team approach is critical to the ultimate success and long-term management of these patients. With proper medical care, children with open NTDs can lead active and productive lives.

For example, in a 20- to 25-year follow-up study of children with open spina bifida who were treated aggressively in a nonselective, prospective manner, these children entered college in the same proportion as the general population and many were actively employed.

CONCLUSION

Preconceptional and antenatal folic acid supplementation can make this a preventable disease.

A thorough assessment of the craniospinal axis needs to be done to look for hydrocephalus, multiple tethering lesions and co-existent cranial malformations, in addition to the apparent defect. Neurological deficits, orthopaedic anomalies, cardiac and gastrointestinal anomalies accompanying the spina bifida defect need to be addressed and managed appropriately.

Significant improvement in motor and sensory function was not seen, but further deterioration in functional ability was curtailed by early surgical intervention. Not all children, but a great majority of patients with spinal dysraphism and hydrocephalus required shunt surgery or CSF diversion. But early correction of defect and regular and vigilant follow up could avoid it in 67% of cases. Long term follow up is needed regardless of initial surgical intervention, as a decline in function is a part of the natural history of this disease and requires early and appropriate diagnosis and treatment.

A multidisciplinary approach is required to address this disease. Early diagnosis, surgical management and rehabilitation can prevent further neurological damage and can improve quality of life in patients with spina bifida.

REFERENCES

- [1] Coran Pediatric Surgery, 7th Edition, Chapter 128, Management of neural tube defects, hydrocephalus, Pages 1674-1676.
- [2] Bruce A. Kaufman, Neural tube defects, Pediatric Clinics of North America, 2004; 51:389-398.
- [3] Vademecum Pediatric Neurosurgery, Chapter 8, Spinal Dysraphism, Pages 143-149.
- [4] Mark S. Dias, MD; Michael Partington, MD, Embryology of Myelomeningocele and Anencephaly, Neurosurgical Focus, 2004; 16(2).
- [5] Toriello HV, Higgins JV. Occurrence of neural tube defects among first-, second-, and third degree relatives of probands: results of a United States study. Am J Medical Genetics, 1983;15:601- 6.
- [6] P. G. SALUJA, The incidence of spina bifida occulta in a historic and a modern London population, J.Anat. ; 1988; 158; 91-93,
- [7] Kumar R. · Singh S.N.; Spinal Dysraphism: Trends in Northern India; J. Pediatric Neurosurgery; 2003;38:133-145.
- [8] Textbook of contemporary neurosurgery, Chapter 25, Spinal and Cranial Dysraphism, Pages 338-340.
- [9] Natalie E. Rintoul, MD; Leslie N. Sutton, MD; A new look at meningocele: Functional level, vertebral level, shunting and the implications in fetal intervention; Official journal of American academy of Pediatrics; pediatric journal;2002;109:3.409
- [10] S. Vande Velde, S. Van Biervliet, et al. ; Achieving Fecal Continence in Patients With Spina Bifida: A Descriptive Cohort Study; The Journal of Urology;12/2007; 17;6:2640-2644.