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CLINICO-PATHOLOGICAL STUDY OF NEURAL TUBE DEFECTS AND ITS SURGICAL MANAGEMENT

Neurosurgery

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ABSTRACT

ObjectiveTo evaluate the clinical profile prognosis and surgical outcome of various neural tubedefects.

Materials and Methods: This is a prospective study of 75 cases of spinal dysraphism managed during a periodof 2 years from December 2013 to December 2015 in Department of Neurosurgery, Bangur Institute of Neurosciences Kolkata. Demographic profiles, clinical presentation of patients with spinal dysraphism, associated hydocephalus, surgical management and outcome were studied.

Results: Out of total 75 cases studied, female patients outnumbered male with 57.33% to 42.66%. Age ranged from 5 days to 40 years and mean age was 2.71 years whereas MMC sac (80%) and paraparesis (66.66%) were frequent mode of presentation. Lumbosacral lesion (40%) was commonest followed by sacral lesion (16%). Hydrocephalus was present in 40% of casesand in 5.3% cases developed hydrocephalus later after repair.

Conclusion: Neural tube defect is debilitating entity and management is challenging. Lump on back and weakness of limb are major factor for children and their parents seek medical service. Lesion in low back (lumbar and lumbosacral) were most common location. Besides repair, majority of them needed CSF diversion surgery for hydocephalus.

Aim of surgical management was to prevent further deterioration, control of hydrocephalus or leak.

KEYWORDS

Neural tube defect; Surgical Management

BACKGROUND

Neural tube defects (NTDs) include all congenital spine and spinal cord defects. These are not limited to just the spinal cord, but include all aspects of the central nervous system. This defect can also affect the future development of the mesenchyme cells, which are undifferentiated mesodermal (middle layer) cells that give rise to such structures as connective tissues, blood, lymphatics, bone, and cartilage. Therefore there are other structural defects that may occur beyond the spinal cord/central nervous system defect. These related defects include but are not limited to bowel and bladder function, the brain, the bones of the spine, and the extremities[1].It is very likely that NTDs occur as a result of multiple genetic and environmental factors[2].As such, descriptions of specific risk factors or genetic components are necessarily complex.

DEFINITIONS

Neural tube defects (NTDs) are a group of birth defects that include various portions of the spine and central nervous system. These defects can be categorized into two groups: open defects and closed defects.

Open defects where the neural tissue is exposed to the air (or amniotic fluid) include spina bifida cystica, which encompasses meningoceles, myeloceles, and myelomeningoceles. Myelomeningocele is a defect in the closure of the neural tube that occurs in the vertebral column. This type of defect can occur anywhere along the spinal column, but is more likely to be placed in the sacral region.

Closed defects in which the skin completely covers the neural tissue, include occult spinal dysraphism, which encompasses diastematomyelia, dorsal dermal sinus, and tight filum terminale. Spina bifida occulta refers strictly to bone fusion defects of the lower spine, generally in the lumbo-sacral region. Anencephaly is a defect in which the head end of the neural tube does not close; this results in the lack of a cranial vault and cerebral hemispheres[2].

Neural tube defects may also occur as part of a syndrome or association. Anorectal malformations are often associated with NTDs, as are defects of the brain. One of these, the Chiari II malformation, includes hindbrain defects as well as hydrocephalus. Hydrocephalus (accumulation of cerebrospinal fluid in the cranium) is often associated with NTDs[1]

MATERIALS AND METHODS:

This is a prospective study of 75 cases of spinal dysraphism managed during a period of 2 years from December 2013 to December 2015 in Department of Neurosurgery, Bangur Institute of Neurosciences Kolkata. Demographic profiles, clinical presentation of patients with spinal dysraphism, associated hydocephalus, surgical management and outcome were studied. We excluded the cases of anencephaly from our study as most of the child with anencephaly are still born or may die in early neonatal period.

RESULTS

Seventy five cases were studied. Mean age at presentation was 2.71 years with age ranging from 19 days to 40 years .Male and female patients were 43% and 57% respectively. The patients were analyzed with respect to their clinical presentation and operative outcome. Mother of 42 patients had received folic acid during antenatal period for a variable period of 2 to 6 months. Fifteen patients were diagnosed to neural tune defect or hydrocephalus by antenatal ultrasound but most of them were diagnosed at 9th month of gestational age.

Neurological presentation

Patients with neural tube defect have protean clinical manifestation. Indivival patients often had more than one symptom or sign. However, one of the clinical features was predominant over the other. Most of the patient with neural tube defect presented with varying degrees of weakness of the limbs (50/75; 66.66%). Graded patchy sensory loss was elicited in 45 cases (60%), while 30 patients (40%) had evidence of sphinter dysfunction. Trophic ulcer was found in 08 patients (10.66%). Five (6.66%) patients presented with back pain.

Cutenous marker

Amongst the skin manifestation, MMC sac over the back was the most common observation, in 42/75 cases (56%). Fifteen (20%) patient had subcutenous lipoma. Hypertrichosis was one of the frequent skin markers, seen in 9 cases (12%). Other skin markers were dermal sinus, cutenous hemangioma and skin tag.

Neuroorthopaedic syndrome

Various orthopaedic deformities were a common accompaniment in neural tube defect patients. The overall incidence of neuroorthopedic syndrome was 51/75 (68%). Often more than one deformity was seen in single patient. Scoliosis was the common neuroorthopedic syndrome seen 20 cases (26%). The other commonest finding was unilateral/bilateral congenital talipes equinovarus, in 20 cases (20%). High arched foot was noted in 5 (6.66%) patients and limb length discrepancy in 5(6.66%). Flat foot was seen in 1(1.33%) case.

Cranial lesion

Following craniospinal MRI, numbers of cranial lesions were evident in association with neural tube defect, among which hydrocephalus was the commonest presentation, seen in 30 patients (40%) most of the patient with required shunt before definitive surgery, as they had clinically apparent raised intracranial pressure. Twenty patients with neural tube defect (26%) had Chiari malformation. Aqueductal

stenosis was noticed in 10 cases (13%). Corpus callosum dysgenesis was found in 8/75 patients with neural tube defect (10.66%).

Level of cranial or spinal defect

The location of defect was variably distributed over the craniospinal aixs. The lumbosacral region of the spine was the commonest site, in 30 patients (40%), followed by the sacral area in 12 (16%). Nine patients (12%) had defect in lumbar region. In 8 patients (10.66%), the defect was noticed in dorsal region. Six patient (8%) had defect in frontonasal region and 5(6.66%) had in cervicodorsal and 5(6.66%) had in occipital region

Operative Results

Patients showed variable results in their clinical paramaters after surgery. Out of 50 patients who had motor weakness of different grades in the preoperative period, only 18 (36%) showed improvement, to varying extents. Five children (10%) had exhibited deterioration in motor strength in the immediate post operative period. The majority of cases (27; 54%) did not manifest any change in the motor function compared to their preoperative status and they remain static postoperatively. Fifteen of 45 patients (33-33%) showed improvement in sensory function, with residual hypoesthesia persisting in most of them. However, in the majority of cases (25/45; 55.55%), sensory hypoesthesia remained the same as before surgery. Only five children showed further deterioration in sensory hypoesthesia in the postoperative period. Of the 30 patients who had bladder dysfunction, only 10 (33.33%) achived subjective improvement in bladder function with an increase in dry periods. In the vast majority of cases (17/30; 56.66%), sensory deficit remains same as in the preoperative period. Six patients with normal preoperative bladder function developed retention of urine in immediate postoperative period, but all of them regained normal bladder function over 1 month following regular clean intermittent catheterisation. Three patients (10%) had deterioration of baldder function. Out of 8 patients who presented with trophic ulcer of the foot, it completely healed in most of them (5; 62.5%). However 2 of the 8 showed only partial response and in one patient it was same as preoperative status. Four patients out of 5 (80%) with back pain showed improvement and in one patient it was same as preoperative status.

Postoperative complication

In the postoperative period, some of the patients developed troublesome complications which required either resurgery or conservative management. Cerebrospinal fluid (CSF) leak from the operative site was the most annoying complication, in 15 patients (20%). Nine of them required reexploration and duraplasty , using fascia lata or synthetic graft., watertight resuturing was all that was needed. Six patients with CSF leak were managed conservatively, by putting them in prone position, pressure dressing and acetazolamide therapy.

Pseudomeningocele, i.e operative site bulge, was noted in 10 patients (13.33%). Five of them required reexploration and duraplasty , using fascia lata or synthetic graft. Five patients with were managed conservatively, by needle aspiration , pressure dressing and acetazolamide therapy.

Operative site infection was another troublesome complication, observed in 10 cases (13.35%). All of them were treated with suitable antibiotics, depending upon culture and sensitivity reports, and they responded well.

Shunt malfunction or infection developed in 10 (13.33%) patients which was managed by antibiotics and shunt removal. Seven out of 10 patients required repeat shunting due to development of subsequent hydrocephalus.

Four (5.33%) cases developed hydrocephalus after repair which was managed by ventriculoperitoneal shunt.

Two patient of large frontonasal encephalocele died on second post operative day

Histopathology

After surgical treatment of various neural tube defects histopahology confirms the clinical and radiological diagnosis as meningomyelocele in 47 cases (62.66) lipomenigomyelocele in 12 cases (16%) encephalocele in 7 cases (9.33%) meningocele in 3 cases (4%) and dermoid in 2 case(2.66%) and for 4 cases histopathology was not available (for 3 split cord malformation and 1 dermal sinus).

DISCUSSION

Neural tube defect is common congenital anomaly of nervous system reported in approximately 1-5 per 1000 live births and the risk of recurrence is 2-3% [3]. More than 95% of cases are contributed by the first affected pregnancies[4]. The incidence varies from country to country and from one geographical region to another. In United States, the prevalence of NTD is 1/1000 live birth. In India the prevalence is 3.63/1000 live births [5].The highest reported from the northern states, namely Punjab, Haryana, Rajasthan and Bihar[6].The incidence of spinal dysraphism is approximately 1 per 1,000 live birth103.This declined 50% between 1970 and 1989 (0.6-1.3 cases per 1000 live births) in the United States[7].Despite aggressive medical care, 10-15% of these children die prior to reaching the first grade. However, most children with isolated myelomeningocele (without major anomalies of other organs) survive to adulthood, and life expectancy is nearly normal [8].

In a study of north India106 Out of 155 cases of spinal dysraphism, 119 had open spina bifida [meningomyelocele (MMC) in 113 (72%), meningocele in 3 (2%) and myelocystocele in 3 (2%)] and 36 had occult spina bifida [split cord malformation on (SCM) without overt MMC sac (pure SCM) in 29 (19%) and midline dermal sinus in 7 (4.5%)]. In a series,most common location on was lumbosacral (44%) followed by dorsolumbar (32%). In this study, lumbosacral region of the spine was the commonest site, in 30 patients (40%), followed by the sacral area in 12 (16%).In this study we found meningomyelocele in 47 cases (62.66) lipomenigomyelocele in 12 cases (16%) encephalocele in 7 cases(9.33%) meningocele in 3 cases (4%) spinal dermoid in 2 cases (2.66%) spilt cord malformation in 3 (4.0%) cases and dermal sinus in 1 (1.33%) case . We excluded the cases of anencephaly from our study as most of the child with anencephaly are still born or may die in early neonatal period. The birth prevalence rate of myelomeningocele was slightly higher in females than in males (1.2:1), based on data from state and national surveillance systems from 1983-1990107. In this study female to male ratio was 1.32:1. The most common presentation on of a closed NTD is an obvious abnormality along the spine such as a fluid-filled cystic mass, area of hypopigmentation or hyperpigmentation , cutis aplasia, congenital dermal sinus, capillary telangiectasia/hemangioma, hairy patch (hypertrichosis), skin appendages [9] .In this study, MMC sac over the back was the most common observation, in 42/75 cases (56%). Fifteen (20%) patient had subcutenous lipoma. Hypertrichosis was one of the frequent skin markers, seen in 9 cases (12%). Other skin markers were dermal sinus, cutenous hemangioma and skin tag.

The brain and spinal cord develop from the neral tube;defective neural tube formation also results in faulty development of brain[10] . Arnold –Chiari malformation, aqueductal stenosis and corpus callosum dysgenesis are common associated brain anomalies present in cases of spinal dysraphism. In this study, we observed Chiari malformation in 26%, aqueductal stenosis in 13%, and corpus callosum dysgenesis in 10.66% of our neural tube defect patient. Earlier studies indicated the presence of Chiari malformation in nearly 90% of all children with MMC. However recent studies suggest that it is clinically significant in 10-20% of these children, especially those aged 3 months or less.[11,12]

The rate of occurance of hydrocephalus in a patient with neural tube defect has been reported over 80% in western literature[13, 14]. In our study, hydrocephalus was found in 30/75 cases of neural tube defect (40%) which is significantly lower than the incidence in the western world. One reason for the low incidence of hydrocephalus may be the fact that being a tertiary referral center, we might be getting selected cases only who had survived their neurological disorder in early phase of life. However other studies [15, 16] from the Indian subcontinent have consistently reported a lower incidence of hydrocephalus (10-12.5%) in cases of neural tube defect.

Various spine and foot deformities are commonly seen in children with neural tube defect. Many series have reported incidence of scoliosis and talipes equinovarus ranging from 30 to 50% [17,18,19]. In our study, the incidence of scolosis and talipes eqinovarus was 26% and 20% respectively.

McLone[20] reported 37% improvement in motor function after surgical

treatment. We observed that 36% of our patients showed improvement, to varying extents. The majority of cases (27; 54%) did not manifest any change in the motor function compared to their preoperative status and they remain static postoperatively. Five children (10%) had exhibited deterioration in motor strength in the immediate post operative period. Fifteen of 45 patients (33-33%) showed improvement in sensory function, with residual hypoesthesia persisting in most of them. However, in the majority of cases (25/45; 55.55%), sensory hypoesthesia remained the same as before surgery. Only five children showed further deterioration in sensory hypoesthesia in the postoperative period.

With a rigorous training program, it is possible to achieve social continence of urine in 75-90% of patients with neural tube defect[21]. However, in our study, only 33.33% of patients regained some sort of subjective control over bladder function leading to an increase in dry periods at least during the daytime. However, the majority of patients (56.66%) showed no alteration from preoperative status. Six patients with normal preoperative bladder function developed retention of urine in immediate postoperative period, but all of them regained normal bladder function over 1 month following regular clean intermittent catheterisation. Three patients (10%) had deterioration of baldder function.

The immediate post operative period in these patients is almost always stormy. Operative site CSF leak, pseudomenigocele and infection are the some of the most troublesome complications which sometimes become difficult to manage. The complications are much higher in open spina bifida, because usually these patients have thin skin which is difficult to oppose and does not heal well. In this study Cerebrospinal fluid (CSF) leak and pseudomeningocele were noticed in 20% and 13.33% respectively. Meticulous dural closure is the only way to prevent these complications, because no other convincing method has been developed to date to overcome these problems of CSF leak and pseudomeningocele.

In a modern era of advanced technology, sophisticated anesthetic techniques, better postoperative care and greater use of antibiotics, the morbidity and mortality of children suffering from neural tube defect has been significantly reduced. McLone[21] reported 2 deathes in series of 100 patients and 1 death from 119 patients was reported by jindal et al.[15], whereas in our series, 2 patients of frontal encephalocele died among 75 cases of neural tube defect. It is evident that surgery performed at an early age helps prevent damage to vital neural tissue. However in india, the majority of patients approach a tertiary center very late because a large number of children are born in rural areas and do not see a neurosurgeon, and most of the time they are operated by general/paediatric surgeons especially in spina bifida aperta.[9,15]

It is logical to state that the entire neuraxis must be screened by craniospinal MRI before definitive surgery to delinate the underlying associated pathologies, because overt Meningocele/ MMCsac/ cutenous markers are only the tip of iceberg, and excision of the Meningocele sac is not the answer to adequate management of these patients.

CONCLUSION

Spinal dysraphism is debilitating entity and management is challenging. Lump on back and weakness of limb are major factor for children and their parents seek medical service. Lesions in low back (lumbar and lumbosacral) were most common locaation. The incidence of hydrocephalus in neural tube dfect is significantly lower in india in comprasion to the western world. Some patients with mild hydrocephalus on scan, but without clinical manifestation, may be followed up without shunt insertion, and in long run, they may not require shunt operation. Besides repair, majority of them needed CSF diversion surgery. Multiple craniospinal anomalies are associated with neural tube defect, making it most complicated developmental anomaly. Multiple tethering lesions coexist in a significant number of cases. Hence, the importance of investigating these patients with craniospinal MRI to screen the entire neuraxis in order to locate lesions other than neural tube defect. Surgical intervention must be done as early as possible to prevent progressive neural tissue damage. Aim of surgical management is to prevent further deterioration and control of hydrocephalus or leak. The surgical microscope assist in defining distorted anatomy and associated pathologies in great detail at the level of spina bifida. Thus, the surgical procedure can be carried out in a single sitting to deal with definite as well as associated pathologies more efficiently in order to achieve a better operative result.

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