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Original Research Paper

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AN OBSERVATIONAL STUDY OF SPECTRUM OF CONGENITAL SPINAL DEFECTS IN CHILDREN ATTENDING PAEDIATRIC OUTPATIENT OF A TERTIARY CARE HOSPITAL IN SOUTHERN INDIA

Dr. Pavithra V — Classified Specialist (Paediatrics), Command Hospital (Western Command) Chandigarh, India

Dr. Arun Tyagi* — Prof & HOD, Department of General Medicine, DVVPF's Medical College, Ahmednagar, Maharashtra *Corresponding Author

ABSTRACT

Congenital anomalies in children are not infrequent and the birth defects of cardiovascular and digestive systems are the most common. Congenital spinal defects have prevalence and incidence of about 2.74% and 1-3 per 1000 live births respectively. The range of spinal defects may vary from a tuft of hair with an underlying spina bifida to various types of spinal dysraphism. This observational study was undertaken to study the spectrum of neural tube defects among children attending the pediatric outpatient of a tertiary care hospital. Twenty-one children were included in the study. Fourteen infants were less than 1-year old and seven were between 2-7 years of age. Fourteen children had meningomyelocele (MMC). The commonest site was in the lumbosacral region. Seven patients of MMC had associated hydrocephalus, and seven had talipes deformity. Other accompanying defects included pes cavus, pectus carinatum, polydactyly and congenital heart defects, seen in one case each. Soft fluctuant swelling over the spine, kyphoscoliosis, flaccid paralysis of lower extremities, and incontinence of urine were the cardinal symptoms. CT Scan and MRI in these children helped us to assess the quantum of deficit involving the vertebra and spinal cord. This paper highlights the range of spinal abnormality seen in children with the similar clinical presentation, and therefore the need for neuroimaging in all cases with suspected neural tube defect (NTD) for proper management and prognostication.

KEYWORDS : Congenital Anomalies, Neural Tube Defect, Folic Acid, Ultrasonography, Alpha-fetoprotein,

INTRODUCTION

Congenital anomalies can be defined as structural or functional anomalies that occur during intrauterine life. These birth defects may be identified before or at birth, or later in life. Approximately 6% of babies are born with a congenital anomaly worldwide. The incidence is higher in underdeveloped and developing countries. The actual figure may be significantly greater as the terminated pregnancies and stillbirths are usually not included in the statistics.[1] Cardiac anomalies, neural tube defects and Down syndrome are the most common severe congenital malformations.[2] Neural tube defects have prevalence rate of about 2.74% and the estimated incidence is about 1-3 per 1000 live births.[3] Females have a 3 to 7 times higher risk compared to males. The incidence is several times higher in China, parts of Africa, Thailand, and India. Overall prevalence in India is 4.5 per 1000 total births.[4] The incidence of neural tube defect (NTD) incidence increases with lower socio-economic status and older maternal age. The recurrence rate in subsequent pregnancies is about 1.5-2.6%.[5]

The causation of congenital spinal defects is multifactorial and may range from genetic to environmental factors. Most open NTDs occur sporadically, but several risk factors may be linked. Some of the known etiologic factors are maternal obesity, gestational diabetes, maternal irradiation, rubella virus infection, folate deficiency, and exposure to antiseizure medicines like valproate and carbamazepine during pregnancy. Known association of NTD exists with chromosomal anomalies like Trisomy 18, trisomy 13, Meckel-gruber syndrome (renal cysts, neural tube defects and polydactyly), and VACTERAL associations .[6]

Additionally, certain teratogenic factors may affect development of more than one organ systems. The congenital spinal defects may be apparent from characteristic symptoms of spinal cord dysfunction or some other external signs. With improved prenatal screening and the advanced imaging techniques, the understanding of various congenital spinal defects has significantly improved.[7] The aim of this study was to analyze various neural tube defects, their clinical features, and association with radiological findings among the children attending pediatric outpatient department (OPD).

MATERIALS AND METHODS

The study was conducted over one-year period, from July 2017 to June 2018, in a hospital in southern India. Twenty-one children (13 females and 08 males), ranging from newborn to 7-year-olds, attending the pediatric OPD with symptoms suggestive of spinal dysraphism, were included in the study. Each child was examined in detail and a thorough evaluation of central nervous system was done. All children were evaluated and subjected to relevant radiological and other relevant investigations. The findings were recorded in a pre-ratified proforma. Two newborns with meningomyelocele had to be discharged on request and were lost to follow up.

RESULTS

Sixteen children presented with meningomyelocele (MMC), two were lost to follow up. The location of MMC was lumbosacral in ten and thoracolumbar in four children. Eight children had flaccid paralysis, and clumsy gait was noted in two. Of these, six had MMC. Six children had incontinence of bladder and bowel, of whom three had MMC. Hydrocephalus was found in seven cases of MMC. Kyphoscoliosis, and sensory deficit was identified in four cases.

Talipes deformity was seen in seven children. Pes cavus, pes planus and heart defect were the other congenital anomalies seen in one child each. Tuft of hair over the spine was seen in two children. Dermal sinus was not seen in any child (Table 1 and 2). The clinico-radiological features of 14 children with MMC is summarized in Table 3.

Six children had neural tube defects other than MMC. The findings among the kids without MMC were as follows-

a. Diastematomyelia with spina bifida L3-S1- 01 ((Split Spinal Cord Malformation (SSCM)
b. Hydromyelia (T8 –T10) with tethered cord with Pes planus- 01
c. Spina bifida with tuft of hair & pes cavus- 01
d. Spina Bifida L5-S3 with kyphoscoliosis- 01
e. Spina bifida L5- S5- 01

All patients were provided requisite comprehensive, care including ventriculoperitoneal (VP) shunt, spinal surgery, physiotherapy, and rehabilitation.

DISCUSSION

Defects in neural folding and neuropore closure in third-fourth week of gestation gives rise to several defects, that may involve dura, skull, and vertebra to variable extent. It is possible to find gross spinal defects without any disruption of skin over the spine (Figure 1).[8] Spina bifida encompasses a wide spectrum of neural tube defects. It is broadly classified as spina bifida aperta and occulta depending upon integrity of skin over open spine. Spinal dysraphism may be associated with various pathological conditions, ranging from simple meningocele with no neural involvement, to myelomeningocele, lipomeningomyelocele, diastematomyelia, myeloschisis and rachischisis leading to wide-ranging neurological deficit (Table 4).[9] With improved prenatal screening, the incidence of Aperta is gradually declining, whereas detection of occulta is increasing with the advent of magnetic resonance imaging.[10] The necessity of thorough radiological work up cannot be overemphasized for management and prognostication of these cases.

Though not as commonly encountered as other pediatric ailments in daily practice like infectious and nutritional diseases, spinal dysraphism remains an important congenital anomaly that requires lifelong care. The true incidence of spinal dysraphism in our country is difficult to quote, as most of the studies are conducted in tertiary care hospitals and thus have an inherent referral bias. A population-based door-to-door survey of mothers living in remote, least developed villages in Uttar Pradesh, India, revealed that the incidence of NTDs was 6·57–8·21 per 1000 livebirths, among the highest in the world. The high numbers of unregistered, untreated cases show that tertiary care center-based studies do not represent the true incidence of NTDs.[11,12]

Meningomyelocoele (MMC) and flaccid paralysis are considered the commonest clinical manifestations of spinal dysraphism, and this was true in our series as well.[13] The development of hydrocephalus is concomitant with MMC and need not be related to the repair of MMC and may develop at any age. Hydrocephalus was observed in 49% of our cases. Hydrocephalus is reported in up to 25% of infants at birth but shunting is required in 80-90% patients as they grow. It may be because aqueductal stenosis, seen in about 80% of children who are likely to develop hydrocephalus at a later stage. [14] Surprisingly, the common association of MMC and aqueductal stenosis was seen in only two of our cases. This conflicting finding could have been owing to the small number of patients in the study. The symptom of bowel and bladder incontinence correlated more with MMC than with other varieties of dysraphism. Bladder and bowel incontinence are commonly associated with patients with spinal dysraphism. Verhoef et al reported 60.9 and 34.1% incidence of urinary and fecal incontinence respective in their series of 179 patients of spina bifida.[15] The clumsy walk, kyphoscoliosis, deformity of foot or leg, disturbance in bladder function alone or in combination was seen in five of our cases who had evidence of tethered cord. The patients with tethered cord may have an array of symptoms, including back and leg pain, progressive motor and sensory deficit, gait disturbances, foot deformity, and more than half have sphincter dysfunction.[16]

There were three cases of SSCM in our study. These cases presented with pyramidal signs and sphincter incontinence. One child had absent ankle reflex. The symptomatology of SSCM depends on the type of malformation. Type I (duplicated dural sac, with common fibrous or osseus midline spur) is seen in about half of cases, is usually symptomatic presenting with scoliosis and tethered cord. Type II (single dural sac containing both hemicords) is milder and the patients may even be asymptomatic. Hypertrichosis is the commonest skin finding (79%) and faun tail is usually observed in SSCM Type I.[17,18]

The treatment of MMC is crucial, and earlier the better, since the pathology adversely affects both, the quality of life (QoL) and longevity. Early detection and complete correction can significantly reduce the neurological disability.[10] Renal damage happens to be the single most frequent cause of morbidity and mortality in patients of MMC. Factors like recurrent and chronic urinary tract infections, urolithiasis usually start damaging kidneys during childhood itself, and evidence of renal dysfunction is found in 30-40% of these children. An important aspect of managing these patients is the prevention and management of progressive renal damage. Also, there is evidence that early diagnosis and neurosurgical intervention may prevent urinary incontinence.[19,20]

Considering the disability, effect on QoL, cost of lifelong care, and reduced life expectancy associated with spinal dysraphism, prevention must be the primary strategy, and all pregnant women must be screened for known risk factors like rubella infection, obesity, gestational diabetes, folate deficiency and medicines. Though several genetic and specific chromosomal disorders are known to be associated with the development of NTDs, such cases account for only a small proportion in live-born infants.[21] Therefore the prevention of modifiable risk factors must be the dictum.

There is compelling evidence that low maternal folate levels are associated with increased incidence of NTD. There is a protective role of pre-conceptional intake of folic acid in reducing occurrence and recurrence of NTD. The analysis of 35 EUROCAT congenital anomaly registry and population serum folate levels by Morris et al revealed that failure to implement mandatory folic acid fortification in the 28 European countries has caused NTDs to occur in almost 1,000 pregnancies every year.[22] Interestingly though, a study by Murphy and Westmark from USA inferred that national food fortification with folic acid was not associated with significant decrease in prevalence of NTDs in the USA.[23] However, food fortification and folic acid supplementation strategies during the periconceptional period is recommended worldwide.[24,25]

In addition to folic acid supplementation, all women must undergo preconceptual counseling on importance of weight and glycemic control, and avoidance of drugs known to be associated with increased risk of NTDs. The preventive strategies must include ultrasonography (USG), and screening of pregnant women with past or family history of birth defects for genetic diseases, maternal serum alfa-fetoprotein (AFP) and MTHFR protein.[26] All pregnant women should be subjected to first- trimester (at 11- 14 weeks) and routine second-trimester (at 18-22 weeks gestation) USG for screening and diagnosis of NTDs. Three-dimensional USG and MRI can be used for evaluation of brain anomalies in selected cases. Invasive prenatal diagnostic methods such as diagnostic amniocentesis for fetal karyotyping and chromosomal microarray may possibly be decided on case-to-case basis.[27,28]

CONCLUSION

The NTDs are among the common congenital anomalies faced by the obstetricians, and pediatricians alike. We have studied the spectrum of NTDs in the population from southern India, and the types of NTDs encountered were generally as described in other studies.We have also reviewed the literature regarding the risk factors and preventive strategies. Folate deficiency is probably the most important modifiable risk factor, and folic acid supplementation is a must for all women during pre-conceptional period. Two key advances related to prevention of NTDs are folic acid fortification of commonly consumed foods, and maternal serum and sonographic screening for prenatal identification of NTDs allowing parents to make informed choices about pregnancy management.

Limitation Of The Study

The limitations of the study are small sample size and incomplete work-up of study subjects because of financial constraints.

Conflict Of Interest- Nil

Table 1: Clinical Features In Cases Of Congenital Spinal Defects

S. No.	Clinical Findings	Number of Patients
1.	Swelling over Spine	14
2.	Large Head/ Hydrocephalus	07
2.	Flaccid Paralysis	06
3.	Incontinence	06
4.	Kyphoscoliosis	04
5.	Talipes deformity	07
6.	Sensory deficit	04
7.	No neurological deficit	04
8.	Pes cavus	01
9.	Pes planus	01
10.	Pectus carinatum	01

Table 2: Radiological Findings In Cases Of Congenital Spinal Defects

S. No.	Radiological Findings	Number of Patients
1.	Spina Bifida Occulta	04
2.	MMC	14
3.	Hydrocephalus	07
4.	Diastematomyelia	03
5.	Tethered cord	03
6.	Lipomeningomyelocoele	02
7.	Aqueductal stenosis	01
8.	Open sacral hiatus S1-S5	01
9.	Hydromyelia T8 –T10	01
10.	Lumbosacral spinal dysraphism with Arnold-Chiari malformation type 1	01

Table 3: Presentation Of Children With Meningomyelocele

S. No.	Clinical/ Radiological Finding	No. of Patients
1.	Flaccid Paralysis	06
2.	Hydrocephalus	07
3.	Talipes Equinovarus	07
4.	Disastetomyelia	02
5.	Tethered Cord	02
6.	Incontinence	03
7.	Aqueductal Stenosis	01
8.	Sensory Deficit	01
9.	Kyphoscoliosis	04
10.	Pectus Carinatum	01
11.	Pes cavus	01

Table 4. Classification Of Spinal Dysraphism[9]

Open Spinal Dysraphism (formerly spina bifida aperta or cystica): occurs when the cord and its covering communicate with the outside; no skin or tissues cover the sac
• myelomeningocele (98% of open spinal dysraphism) • myelocele • Hemimyelomeningocele • hemimyelocele
Closed Spinal Dysraphism (formerly spina bifida occulta): occurs when the cord is covered by other normal mesenchymal elements
• with subcutaneous mass lipoma with dural defect lipomyelomeningocele lipomyelocele terminal myelocystocele meningocele limited dorsal myeloschisis
• without subcutaneous mass posterior spina bifida (isolated defect of the posterior neural arch of vertebra) intradural lipoma filar lipoma tight filum terminale persistent terminal ventricle
• disorders of midline notochordal integration dorsal dermal sinus dorsal enteric fistula neurenteric cyst split cord malformations diastematomyelia diplomyelia • disorders of notochordal formation caudal regression syndrome type 1 type 2 segmental spinal dysgenesis

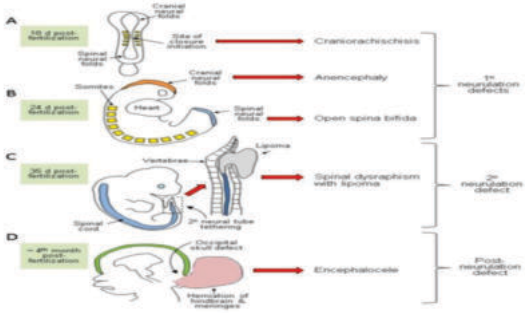

Figure 1: Diagrammatic Representation Of The Developmental Origin Of Malformations Broadly Classified As Neural Tube Defects In Humans.[8]

(a,b) Disorders of primary neurulation include craniorachischisis (a) in which the neural tube fails to initiate closure, leaving most of the brain and the entire spine open. If closure initiates successfully, then the cranial and/or spinal neural folds may fail to close (b) generating exencephaly/ anencephaly and open spina bifida (myelomeningocele), respectively. (c) Disorders of secondary neurulation comprise failure of the neural tube to separate completely from adjacent tissues, resulting in tethering and diminished mobility.

The spinal cord is covered by skin and often associated with fatty tissue accumulation (lipoma) through as-yet-unknown mechanisms. (d) Postneurulation defects can arise when the bony structure of the skeleton fails to develop fully. Herniation of the meninges, with or without brain tissue, through a skull defect (shown here as occipital but sometimes parietal or fronto-ethmodial) generates encephalocele, while an analogous defect in the spinal region produces meningocele.

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