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Pathological Patterns and Clinical Presentations of Spinal Dysraphism in Different Age Groups

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ABSTRACT

Background: Spinal dysraphism (SD) results from failure of fusion of the caudal neural tube. Its incidence is 2-4 / 1000 live births. **Objective:** To study the pathological patterns and clinical presentations of spinal dysraphism and its association with some known predisposing factors of congenital disorders. **Study Design:** Descriptive cross-sectional study. **Settings:** Department of Neurosurgery, Dow University of Health Sciences and Civil Hospital, Karachi Pakistan. **Duration:** Six years from October 2015 to September 2018. **Methods:** This study was conducted in 100 consecutive patients, who were diagnosed as cases of spinal dysraphism based on history, clinical examination, and MRI appearances. **Results:** Large majority of patients (64 out of 100 or 64%) were in the first year of life. The most common clinical presentations were muscular weakness (70%), foot deformity (40%), bladder disturbance (36%) and skin abnormality (28%). Myelomeningocele was the most common pathological pattern (56%) followed by lipomyelomeningocele (16%), diastematomyelia (10%), congenital dermal sinus (8%), hypertrophied filum terminale (8%) and meningocele (2%). Most patients came from a lower socioeconomic stratum and there was no association with consanguinity and maternal age and only two patients had affected siblings. **Conclusion:** Knowledge regarding different types of spinal dysraphism and their presentations is essential for all medical practitioners.

Keywords: Spinal dysraphism, Neural tube defects..

INTRODUCTION

Spinal dysraphism (SD) is a term that describes a group of congenital spinal anomalies, resulting from failure of closure or defective closure of the neural tube early in fetal life.[1] The common feature of the group is an abnormal development of the midline structures of the back, with absence of some of the neural arches which may be associated with external stigmas like pigmentation, tuft of hair or sinus and/or defects of filum terminale, nerves, and spinal cord.[2]

Based on the type of spinal defect the condition is classified into two groups; spina bifida occulta, the closed variety, consisting of a defect in the vertebral arches with no externally visible sac on the back.[3] This group includes the incidental finding of bifid spinous processes and laminae without neurological involvement, seen in 5-10 % of general population,[4] intradural lipoma, tethered cord due to hypertrophied filum terminale and diastematomyelia with splitting of spinal canal and cord by a bony spur.[5] In spina bifida cystic or aperta, the open type, the vertebral defect is associated with a cystic mass on the back, with evident involvement of neural tissue, myelomeningocele, which may be associated with a lipomatous mass, lipomyelomeningocele and with CSF containing dural sac without neural tissue, meningocele.[6]

The basic defect of spinal dysraphism starts in the first 8 weeks of fetal life.[7] The neural tube which develops from folding of ectodermal neural plate is separated from the ectoderm of the skin by incorporation of the mesoderm which forms the bony elements, meninges, and muscle.[8] Failure of fusion of neural tube in the midline results in, posterior spinal abnormalities such as myelomeningoceles occurs.[9] Incomplete separation of ectoderm from the neural tube leads to cord tethering, diastematomyelia, or a dermal sinus, while premature separation of the cutaneous ectoderm from the neural tube with incorporation of mesenchymal elements between the two results in the development of lipomas.[10]

Of the many factors implicated in the development of a spinal dysraphism, genetic, racial, and environmental

factors are the most important.[11] In most infants with dysraphism there is no history of previously affected children in the family. However, families with one affected child are at increased risk of having offspring with neural tube defects than children without affected siblings.[12] The risk is 1 in 20-30 for subsequent pregnancies, and if 2 children are affected, the risk becomes 1 in 2.[13] Insulin-dependent diabetes mellitus, intrauterine exposure to anticonvulsants like valproate, carbamazepine and drugs to induce ovulation (1.5%) have all been implicated as maternal risk factors.[14]

Studies in the early 90s showed that fifty percent of neural tube defects are due to nutritional deficiency of folic acid and are preventable. Correction of folic acid deficiency is said to be an effective means of primary and recurrent prevention.

There are significant racial and geographic variations and the incidence of open neural tube defects vary widely not only among countries but also among different regions within the countries. In multiracial societies, the disease is 2.5 times more frequent in whites than blacks.[15]

Although most cases of spinal dysraphism are diagnosed at birth, some are not apparent in early life and present with progressive neurologic deterioration as the child grows.[16]

Since most varieties or at least the close forms are compatible with normal life expectancy it is essential that these be diagnosed early and managed appropriately before irreversible neurological damage sets in.[17] Data regarding the relative distribution of various types of spinal dysraphism in the Pakistani population and their clinical presentation does not exist. To ascertain this relative distribution of the lesions, their clinical presentation in our population, and their association with known causative factors for congenital disorders, the following study was conducted at the Department of Neurosurgery, Dow University of Health Sciences and Civil Hospital, Karachi. This unit is part of the largest tertiary care teaching hospital of Pakistan and receives many neurosurgical patients from southern Sindh and Baluchistan provinces including those with congenital disorders like spinal dysraphism.

The objectives of this study were:

a. To describe the pathological patterns and clinical presentations of spinal dysraphism in different age groups.

b. To ascertain if there is the association with certain other causative factors of congenital disorders such as maternal age, consanguinity of parents, involvement of other siblings, socioeconomic factors, intrauterine drug exposure and exposure to other illnesses

METHODS

This was a cross sectional study conducted at Department of Neurosurgery, Dow University of Health Sciences and Civil Hospital Karachi from October, 2015 to September, 2018. The study was done on 100 consecutive cases of spinal dysraphism which were selected by non-probability purposive sampling technique. Cases of all ages and of both sexes proven to be of spinal dysraphism based on clinical and MRI findings were included. Patients with spinal stenosis, prolapsed inter-vertebral disc, spinal trauma, spinal neoplasm and operated cases of spinal dysraphism were excluded. Patients were admitted through outpatient or emergency department at Neurosurgery Department, Civil Hospital Karachi. Informed consent was taken from the patient or guardian. Detailed history of patient was taken to determine the age, duration of symptoms and any previous treatment. Complete and thorough clinical examination was performed to ascertain the neurological status. Patients with obvious myelomeningocele and not candidates for surgery were not investigated with MRI. In all other cases MRI scan of the involved region was performed. Diagnosis of Spinal dysraphism was based on history of the patients, clinical features and MRI appearances. MRI scan findings were interpreted by consultant radiologist. Confounding variables were controlled by strictly following the inclusion and exclusion criteria and also by controlling assessors' proficiency. Patients less than one year of age also had ultrasound examination of the head for ventricular size. All patients were clinically assessed to exclude other congenital disorders. Where clinically indicated further investigations were performed.

The whole data was collected by me on a proforma specially designed for this study.

The data was entered and analyzed on SPSS (Statistical program for Social Sciences) version 16.

RESULTS

Between October 2015 to September, 2018 a total of 100 cases of spinal dysraphism were enrolled after taking written consent. Out of these 48 (48%) patients were male and 52 (52%) females. Their ages ranged from 5 days to 25 years (mean age = 4.31 year $\pm$ 6.68). A large majority of patients (64 out of 100 or 64%) were in the first year of life (Table 1).

The most common presentations were muscular weakness (70%), foot deformity (40%), bladder disturbance (36%) and skin abnormality (28%). Myelomeningocele (56%) was the most common pathological pattern followed by lipomyelomeningocele (16%), diastematomyelia (10%), congenital dermal sinus (8%), hypertrophied filum terminale (8%) and meningocele (2%) (Tables 2 and 3).

Table 1: Age distribution of patients (n=100)

Age	Male	Female	Total	Percent
> 0 – 1 year	32	32	64	64
> 1 – 5 years	4	2	6	6
> 5 – 16 years	8	14	22	22
> 16 years	4	4	8	8
Total	**48**	**52**	**100**	**100**

(Age range: from 5 days to 25 years, Mean age: 4.31 years, Std. Deviation: 6.68years).

Table 2: Frequency of clinical features of patients (n= 100)

Clinical presentation	Frequency	Percent
Muscular weakness	70	70
Foot deformity	40	40
Bladder disturbance	36	36
Skin stigmata	28	28
Gait abnormality	16	16
Scoliosis	16	16
Back pain	16	16
Sensory abnormality	12	12
Bowel disturbance	12	12

Table 3: Frequency of pathological patterns (n=100)

Pathological Pattern	Male	Female	Total	Percent
Myelomeningocele	30	26	56	56
Lipomyelomeningocele	8	8	16	16
Diastematomyelia	4	6	10	10
Congenital dermal sinus	4	4	8	8
Hypertrophied filum terminale	2	6	8	8
Meningocele	0	2	2	2
Total	**48**	**52**	**100**	**100**

Of the 64 patients in less than 1 year of age, 52 patients (81.3%) had Myelomeningocele, 10 (15.6%) had lipomeningocele and 2 had meningocele. Six patients were in age group between 1-5 years, 4 of whom had myelomeningocele and 2 lipomeningocele. Of the 22 patients between 5-16 years of age, 10 had diastematomyelia, 7 had tethered cord due to hypertrophied filum terminale, 4 had lipomeningocele and presented with congenital dermal sinus. Eight patients presented late (after 15 years of age), 7 of whom had congenital dermal sinus and one had tethered cord due to hypertrophied filum terminale. (Table 4)

Of the hundred patients, 12 were born of consanguineous marriage (between first cousins). There were no affected siblings of spinal dysraphism in the group. None of the mothers were insulin dependent diabetic or had history of anticonvulsants intake. Sixteen out of hundred patients were born with mothers having age more than 30 years.

Table 4: Age wise distribution of pathology (n=100)

Pathological Pattern	< 1 year	> 1-5 years	> 5-16 years	> 16 years
Myelomeningocele	52	4	-	-
Lipomyelomeningocele	10	2	4	-
Diastematomyelia	-	-	10	-
Congenital dermal sinus	-	-	1	7
Hypertrophied filum terminale	-	-	7	1
Meningocele	2	-	-	-
Total	**64**	**6**	**22**	**8**

DISCUSSION

Spinal dysraphism is a common malformation of the nervous system reported in 200-400 / 100,000 live births.[18] As open spinal dysraphism is apparent at birth, these patients present to the pediatrician and the neurosurgeon within a few days of birth or sometimes referred by obstetrician before birth. In Pakistan large numbers of deliveries are conducted at rural areas, at home or at primary health centre (PHC) and these patients may not quickly reach the tertiary care hospitals. Occult spinal dysraphism is associated with enuresis, lumbar trichosis, lumbar discoloration, extremity anomalies and gait disturbances.[19] These lesions are important for all physicians to recognize as prophylactic surgery has a high likelihood of changing the natural history in which a gradual loss of function may be replaced by clinical stability.[20] Although the disease in many patients is recognized in infancy and childhood because of cutaneous signs, at least one third of patients may not have apparent skin changes.[21]

The onset of signs and symptoms of occult spinal dysraphism may be so gradual that a patient's initial presentation to a neurosurgeon may not be until adulthood.[22] It is for all of these reasons that it is important for the medical community to be aware of this complex and fascinating group of problems.

In our study the age of the patients ranged from 5 days to 25 years with mean age of 4.31 years. 64% of the patients were below one year, 6% were between 1-5 years, 22% were between 5-16 years and 8% were above 16 years (Table-1). It is comparable to the study conducted by Anupum Jindal, Ashok Kumar and Raj Kamal.[23] They retrospectively analyzed 119 patients of spinal dysraphism. The age of the patients varied from 8 days to 47 years with mean of 7.2 years. 18% of the patients were below one year, 35% were between 1-5 years, 36% were between 5-16 years and 11% were above 16 years. There were 60% male and 40% female in the study while in our study the males and females were 48% and 52% respectively.

In our study the most common clinical presentation was muscular weakness (70%) followed by foot deformity (40%), bladder dysfunction (36%), skin stigmata (28%), gait abnormality, scoliosis, back pain (16% each) sensory abnormality and bowel disturbance (12% each) [Table-2]. In the study by A Jindal *et al* they also found muscular weakness as the most common symptom (75%) followed by gait abnormality (50%), loss of sensations (40%) and bladder and bowel disturbances (30%). In another study of 78 patients of spinal dysraphism by C.K. Chang, et al,[24] the most common clinical presentation was bladder and bowel disturbances (73% and 70% respectively) followed by foot deformity (63%) and scoliosis (28%).

The most common pathological pattern in our study was myelomeningocele (56%) followed by lipomyelomeningocele (16%), diastematomyelia (10%), congenital dermal sinus tract (8%), hypertrophied filum terminale (8%) and meningocele (2%). There are various studies having comparable statistics.[25,26,27]

As would we expected, myelomeningocele was the commonest dysraphic disorder in the first year as it comes to the notice immediately after birth due to the obvious defect and gross neurological deficit. There were only six patients between 1-5 years of age as large majority of patients has already been diagnosed during the first year.[28] There was a second peak of 22 patients between 5-16 years of age. These were the patients who had lesions which are not obvious at birth (diastematomyelia and tethered cord due to hypertrophied filum terminale) but manifest themselves with prepubertal growth spurt.[29]

Despite the known risk of familial involvement in dysraphism none of the patients in this study were found to have affected siblings. The consanguinity rate was 12% which is probably the rate of consanguinity in marriage in our population.

CONCLUSION

Spinal dysraphism is a condition which often compatible with normal life span. Moreover, if the deformities are not picked early and corrected, this condition may lead to lifelong disability. Knowledge regarding different types of spinal dysraphism and their presentation is essential for all medical practitioners.

LIMITATIONS

The type of study is a cross sectional study, which traditionally can only establish association and not causality. The study is also a single center study in a large urban area, had the study been conducted at a national level the incidence rate might have been different. We also did not conduct any genetic analysis of the patients to include a genetic basis of SD.

SUGGESTIONS / RECOMMENDATIONS

Further studies including multicenter data and genetic analysis may reveal more information regarding SD.

CONFLICT OF INTEREST / DISCLOSURE

None to declare at the time of the publication.

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