

Pattern of Presentation of Spinal Dysraphism: A Study of 72 Patients in Hayatabad Medical Complex Peshawar, Pakistan

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DOI: 10.36552/pjns.v24i3.433

ABSTRACT

Objective: To review the pattern of presentation and current understanding of patients with spinal dysraphism in our local population.

Material and Methods: Cases of spinal dysraphism of any gender and age were admitted via OPD, emergency or referred from another department were included in the study. Information on demographics, developmental history, presenting symptoms, presence/absence of back swelling, hairy patches, a nevus, dimple, an appendage/skin tag, lower limb function, sensory/motor deficit, bowel and bladder dysfunction were recorded. MRI spine was done in all patients to know the exact diagnosis.

Results: Out of 72 cases, 52 (72.2%) presented with spina bifida Aperta (spina bifida cystica) while 20 (27.7%) with spina bifida occulta. Total 53 (73.6%) patients presented at the age of 0 – 1 years. 41 (56.9%) of the patient presented with visible sac, 35 (48.6%) swelling over the back, in 5 (6.9%) of patient have hairy patch and dermal sinus each, while 28 (38.8%) patients have neurological deficit. Most common type of spinal dysraphism was myelomeningocele 45 (62.5%). Postoperative course of patients with spinal dysraphism was found to be uneventful in 56 (77.7%), wound infection was seen in 11 (15.2%), deterioration of neurological status in 3 (4.16%) of cases.

Conclusion: Spinal dysraphism is not an uncommon condition in our local population its clinical presentation and features are in line with internationally reported literature. Our population is least aware of the adverse neurological outcomes of the condition and face difficulties to access the adequate healthcare for spinal dysraphism.

Keywords: Spina bifida, myelomeningocele, spinal dysraphism.

Abbreviations: SCM: Split Cord Malformation. HCP: Hydrocephalus. ACM: Arnold Chiari Malformation. TCS: Tethered Cord Syndrome. CT: Computed Tomography. CSF: Cerebrospinal Fluid. MRI: Magnetic Resonance Imaging.

INTRODUCTION

Spinal dysraphism is a group of congenital deformities of the spine in which the dorsal midline structure fails to fuse in normal fashion. It can be broadly classified into spina bifida aperta (open type also named spina bifida aperta) in which overlying skin is not intact causing leakage of cerebrospinal fluid. The examples

are myelomeningocele, myeloschisis. The second type is Spina bifida occulta (close type) in which the defect is fully covered with skin and the examples are meningocele, lipomyelomeningocele, split cord malformation and congenital dermal sinus.¹⁻³ The incidence of spinal dysraphism is 1-3/1000 live births.⁴

In spinal dysraphism, the primary embryological

defect is due to failure of, or incomplete closure of neural tube. This process normally occurs during third to fourth week of intrauterine life. The exact etiology is not known, however genetic, nutritional and environmental factors are involved in the development of this anomaly.^{5,6}

Spinal dysraphism patients present with swelling in the back region which is often noticed at birth. The skin over the swelling may be ruptured during labour resulting in cerebrospinal fluid leak, contamination and meningitis.⁷ In Spina bifida occulta (close type), one can see hairy patches, dimples, a nevus, an appendage or skin tag. Some older children may present with sensory motor and sphincter dysfunction depending upon the severity and level of anomaly.⁸ Defect predominantly involves the thoracolumbar, lumbosacral, lumbar, thoracic, cervical areas in decreasing order of frequency. The associated anomalies are hydrocephalus, chiari malformation, skeletal abnormalities such as kyphosis, scoliosis and deformity of long bone and feet.^{9,10}

A meticulous examination of the nervous system is mandatory to diagnose the case of spinal dysraphism. Initially a radiological assessment should be done keeping in view the clinical condition. Plain x-ray reveals the skull defect, spine deformity and bony anomalies.¹¹ Ultrasonography is done for assessment of hydrocephalus. MRI is the gold standard radiological modality to study the neural tissue anomalies and to ascertain the degree of chiari malformation and hydrocephalus¹².

The management of patients with spinal dysraphism is mostly surgical and often need multidisciplinary approach. The mortality rate has been found to be 35 – 45% in first year and 50 – 60% in first 3 – 5 years.¹³ Parents should be counselled regarding the prognosis and different treatment strategies.

We live in under developed part of the world where nutritional status of potential mothers is not up to the mark hence hypothetically higher risk of spinal dysraphism. There is little literature available on patterns of spinal dysraphism in our local population. This study is primarily designed to elaborate the various patterns of this problem in our population and relative sub-share of each secondary-type of spinal dysraphism which will help the health authorities to plan community sensitization and awareness programs. Spina bifida occulta go unnoticed in our community until late growing ages and patients

develop permanent neurologic deficit. It is our opinion that community awareness based on burden of disease of its various presentations may help masses seek timely medical help.

MATERIAL AND METHODS

Study Design & Setting

This descriptive cross-sectional study was done in Neurosurgery department, Hayatabad Medical Complex, Peshawar from February 2018 to March 2020.

Inclusion Criteria

Cases of spinal dysraphism of any gender and age range from 0 – 18 years were admitted via outpatient department, emergency or referred from pediatric/Obstetric department were included in the study.

Data Collection

The data was collected on predesigned proforma. Informed, written consent was taken from parents of all children regarding their inclusion in the study. The institute ethical committee for research evaluation was approached for approval of the proposed study.

A total of 72 patients were recruited in this study. Data was collected about demographics, developmental history, presenting symptoms, duration of symptoms, birth history and maternal history.

Clinical Examination

A detailed clinical examination were performed and findings recorded like presence/absence of back swelling, hairy patches, a nevus, dimple, an appendage / skin tag, lower limb function, sensory deficit, motor deficit, bowel and bladder dysfunction, size and shape of head, skeletal abnormality like kyphosis, scoliosis and deformity of long bone and feet were also recorded.

MRI spine was done in all patients to know the exact diagnosis. CT scan spine was done in selected patient to detect possible associated anomaly like, split cord malformation (SCM), Hydrocephalus (HCP), Arnold Chiari Malformation (ACM), and tethered cord syndrome (TCS). Radiological finding were recorded for diagnosis and the level of dysraphism. Postoperatively, the patients were followed up to three months.

Data Analysis

Immediate complications were recorded. SPSS 20.0 was used for the analysis of data and presented in tabular form.

RESULTS

Background Information

The sample size was 72, all these cases were diagnosed with spinal dysraphism from February 2018 to March 2020. Out of 72 patients 52 (72.2%) presented with spina bifida aperta, while 20 (27.7%) with spina bifida occulta.

Age Incidence

A total of 53 (73.6%) patients presented at the age of 0 – 1 years, 9 (12.5%) between 2 – 6 years, 5 (6.9%) between 6 – 12 years and 5 (6.9%) 13 – 18 years of age.

Gender Distribution

Male were 48 (66.6%) and female 24 (33.3%) with male to female ratio was 2:1.

Table 1: Age Wise Distribution (n = 72).

Age	Spina Bifida Aperta	Spina Bifida Occulta	Percentage
< 24 Hrs	8	2	13.88%
2 Days to 1 Month	18	5	31.9%
2 Months to 01 Year	15	5	27.7%
2 Years to 6 Years	5	4	12.5%
6 Years to 12 Years	3	2	6.94%
13 Years to 18 Years	3	2	6.94%

Clinical Presentation

41 (56.9%) of the patients presented with visible sac, 35 (48.6%) with swelling over the back, in 5 (6.9%) of patient we observed a hairy patch and dermal sinus each while 28 (38.8%) patient had neurological deficit. Out of 28 patients, 16 (57.14%) were having paraplegia 12 (42.8%) having grade 2 or less

paraparesis while 15 (53.7%) presented with a combination of both lower limb weakness and bowel/bladder dysfunction.

In our study the most common type of spinal dysraphism was myelomeningocele 45 (62.5%), followed by lipomyelomeningocele 12 (16.6%), dermal sinus 4 (5.5%), Klippel – Feil syndrome or blocked vertebrae 2 (2.7%), tethered cord syndrome 6 (8.3%) and split cord malformation seen in 3 (4.16%). Myelomeningocele was mostly associated with hydrocephalus either before or after surgical repair which later need a CSF diversion procedure

In our study, the most common site was lumbosacral 20 (27.7%), followed by dorsolumbar 18 (25%), followed by lumbar 15 (20.83%) area. Only 3 (4.16%) had cervical meningocele.

Postoperative course of patients with spinal dysraphism was found to be uneventful in 56 (77.7%), wound infection was seen in 11 (15.2%), deterioration of neurological status in 3 (4.16%) and re exploration was done in 2 (2.77%) of cases.

Table 2: Modes of Clinical Presentation (n = 72).

Clinical Presentation	No. of Patients
Visible sac	41
Swelling over back	35
Dermal sinus	5
Hairy patch	5
Neurodeficit	28

Note: patient may present with one or more than one clinical presentation.

Table 3: Distribution of Cases Based on Type of Spinal Dysraphism (n = 72).

Type	No. of Patients	Percentage
Myelomeningocele	45	62.5%
Lipomyelomeningocele	12	16.6%
Dermal sinus	4	5.5%
Klippel – Feil Syndrome	2	2.7%
Tethered Cord Syndrome	6	8.3%
Split Cord Malformation	3	4.16%

Table 4: Site of Spinal Dysraphism (n = 72).

Site	No. of Patients	Percentage
Cervical	3	4.16%
dorsal	11	15.27%
Lumbar	15	20.83%
Dorsolumbar	18	25%
Lumbosacral	20	27.7%
Sacral	5	9.72%

DISCUSSION

The estimated incidence of spinal dysraphism worldwide is about 1-3/1000 live births. It seems that the prevalence rate has dropped all over the world due to the better antenatal care, folic acid supplementation and good nutrition for women.¹⁰ During 3rd to 4th week of fetal life the primary neural tube fail to close leading to spinal dysraphism, which is broadly classified in to spina bifida Aperta (open type) in which overlying skin is not intact and spina bifida occulta (close type) in which the overlying skin is intact but the spinal cord is anchored to various tissue like skin, subcutaneous tissue, adipose tissue or cartilage.¹⁴

In our study the most common age at presentation was 0 – 1 year in 53 (73%) of patients. A similar study was done by Mahapatra and Meneze and showed that 70% and 76% patients respectively, were presented in pediatric age group.¹⁵ We observed a male predominance 48 (66%), which coincide with a study done by Raj and Singh in Northern India.¹⁶

In our study, 41 (56.9%) of the patient presented with visible sac, 35 (48.6%) with swelling over the back, in 5 (6.9%) of patient we observed a hairy patch and dermal sinus each while 28 (38.8%) patient had neurological deficit. Similar results were observed in a study done by Venkatesh in India in 2016 showing 72% of patients presented with a visible sac over the back and a minority presented with dermal sinus and neurological deficit.¹⁷

In our study the most common type of spinal dysraphism was myelomeningocele 45 (62.5%), followed by lipomyelomeningocele 12 (16.6%), dermal sinus 4 (5.5%), Klippel – Feil syndrome or blocked vertebrae 2 (2.7%), tethered cord syndrome 6 (8.3%) and split cord malformation seen in 3 (4.16%). Myelomeningocele are mostly associated with

hydrocephalus either before or after surgical repair which later need a CSF diversion procedure. A similar study was done by Pornswan et al in 2005 and Ahmed et al in 2010.^{18,19}

Spinal dysraphism commonly occur at lumbosacral area, many study reported lumbosacral area as common site, however Asindi and Al-Sehri had conducted a contradictory report showing dorsolumbar area the most common site.²⁰

Postoperative course of patients with spinal dysraphism was found to be uneventful in 56 (77.7%), wound infection was seen in 11 (15.2%), deterioration of neurological status in 3 (4.16%) and re exploration was done in 2 (2.77%) of cases. A similar study was done by Galhom et al showing a dramatic improvement in symptoms with only a minority of patients experience wound infection and worsening of neuro deficit.²¹⁻²³

CONCLUSION

Spinal dysraphism is not an uncommon condition in our local population its clinical presentation and features are in line with internationally reported literature. Our population is least aware of the adverse neurological outcomes of the condition and face difficulties to access the adequate healthcare for spinal dysraphism.

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Additional Information

Disclosures: Authors report no conflict of interest.

Ethical Review Board Approval: The study was conformed to the ethical review board requirements.

Human Subjects: Consent was obtained by all patients/participants in this study.

Conflicts of Interest:

In compliance with the ICMJE uniform disclosure form, all authors declare the following:

Financial Relationships: All authors have declared that they have no financial relationships at present or within the previous three years with any organizations that might have an interest in the submitted work.

Other Relationships: All authors have declared that there are no other relationships or activities that could appear to have influenced the submitted work.

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Sr.#	Author's Full Name	Intellectual Contribution to Paper in Terms of:
1.	Sohail Amir	Study design and methodology.
2.	M. Nawaz Khan	Paper writing, referencing, data calculations and Correspondence
3.	M. Imran	Data collection and calculations
4.	Ayaz Afridi	Analysis of data and interpretation of results etc.
5.	M. Atif	Literature review and manuscript writing
6.	Mushtaq	Analysis of data and quality insurer
7.	Shahid Ayub	Proof reading

Date of Submission: 11-04-2020

Date of Revision: 27-8-2020

Date of Online Publishing: 25-09-2020

Date of Print: 30-09-2020