



# Spinal Dysraphism: Presentation, Management and Outcome of Patients undergoing Surgery

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## Abstract

The development of the spinal cord can be summed up in three developmental stages: gastrulation (2–3 weeks), primary neurulation (3–4 weeks) and secondary neurulation (5–6 weeks). Any sort of deviance in any of these stages causes anomalies in spine and spinal cord; these anomalies are referred as spinal dysraphism. The approximated incidence of spinal dysraphism is about 1/1000 live births. The present study aims at varied presentations, management and surgical consequences in spinal dysraphism. Methods: A study on surgical treatment of spinal dysraphism was done over the period of 6 months from December 2019- May 2020 at GMC, Jammu. In the study at hand; the clinical aspects, management and surgical outcomes in case of spinal dysraphism were characterized. Results: A total of 30 patients of spinal dysraphism were operated with 70% males and 30% females. The age group selected for the study was between 0-3 years. In early post-operative period the most common symptoms were found to be meningitis (6%), minor wound infection (10%), major wound infections (14%) and 14% patients reported CSF leakage. Out of 30 patients who underwent surgery, 67% patients had good outcome, 20% reported satisfactory outcome and 13% reported poor outcome with an overall mortality rate of 13%. Conclusion: Spinal dysraphism is a complex disorder and requires a multidisciplinary approach for management. The present study suggests that surgery is the prominent treatment along with good post-operative care for the better life of the patient.

**Keywords:** Spinal dysraphism, Myelomeningocele, surgery

## Introduction

Spinal dysraphism postulates an array of congenital abnormalities leading to a defective neural arch along which the meninges are herniated causing various clinical manifestations (Warner et al., 2019). These malformations of the spine and the spinal cord occur between the third and fourth week of gestation (Trapp et al., 2021). The term spinal dysraphism conclude the set of defects inferred from the abnormal development of ectoderm, mesoderm and endoderm tissues and its outcome might affect brain, extremities, bones, bladder and bowel functions. These de-formalities are classified into aperta (visible lesion) and occulta (with no external lesion) (Premlal et al., 2018). The calculated rate of incidence of the disease is 1-3/1000 live births. The disease is found to be more prevalent in female newborns 1.0–1.7 than male, 1.0 (Venkataramana, 2011).

Spinal dysraphism arise from a very early event in the embryonic development. Around third week of the developmental stage, the neural plate folds up to form neural tube. The crown of the neural tube forms the head and the remaining tube develops into the spinal cord and the spine (Eagles and Gupta, 2020). Spinal dysraphism occurs when a segment of the neural tube, to be

developed into the spine doesn't close completely (Blount et al., 2019). Depending on the anomalies caused in the spinal cord, the spinal dysraphism is divided into many types such as myelomeningocele (spina bifida), diastematomyelia (split cord malformation), lipomyelomeningocele (spinal cord lipoma), tight filum terminale, dermal sinus tract and tethered spinal cord (Aurel and Livii, 2020).

Of all the types, Myelomeningoceles or the spina bifida is the most common type of spinal dysraphic conditions impacting the lower urinary tract (Thompson, 2014). The term spina bifida comes from the latin, meaning, open spine. The incidence of spina bifida is approximately 1 in 1200 to 1400 births (Ifitikhar and De Jesus, 2020). It is usually associated with skin defects with CSF leak risks. The open spina bifida is also the most severe form of spinal dysraphism. The condition is associated with poor ability to walk, impaired bowel and bladder conditions and hydrocephalus (accumulation of fluids in brain). The majority of the kids with spina bifida have normal intelligence. Though there are reports of children affected with spina bifida having learning disability (Sims-Williams et al., 2017).

Spinal dysraphia occulta are the closed form of spinal dysraphism. In this form, the skin forms a layer over the neural

tissues. In majority of the cases, a skin marker is found to be present (Dias and Rizk, 2019). The recognition of these markers is essential to understand the type of dysraphism. These cutaneous marks can induce a spinal cord injury leading to progressive or sudden neurological deterioration. These marks can be in the form of hairy patch, an appendage, skin tag, cutaneous nevus, small dimple with a pinhole or a lipoma. Occult spinal dysraphism can be of varied embryologic etiologies but mostly they are associated with tethering of the spinal cord. The most common occulta forms are lipomas, diastematomyelia, diplomyelia, split cord malformations and dermoid tumors (Bradko et al., 2019).

For the diagnosis of the disease in utero, prenatal testing for neurological anomalies can be done using ultrasound or by maternal Alpha Feto Protein (AFP) screening. It has been reported that females bearing fetus with spina bifida has elevated levels of AFP and reduced levels of Human chorionic gonadotropin (HCG) and un-conjugated estriol (Reghunath et al., 2019). The maternal serum testing can detect 80% cases of spina bifida. Whereas, sonography can detect upto 90% cases of spina bifida. The testing for in utero defects should be done at 12, 22 and 32 weeks. The maternal periconceptional intake of folic acid has been known to reduce the incidence of spina bifida (Gotha et al., 2020). Other strategies to investigate spina bifida are by Magnetic Resonance Imaging (MRI), Doppler study of brain for resistive index of vessel to check for hydrocephalus and Chiari malformation, Computed Tomography (CT) to check for hydrocephalus, the maternal serum testing for chromosomal anomalies is also used (Szopa et al., 2020).

The children affected with spinal dysraphism reported to have a greater risk for showing worse levels of internalizing symptoms and low self esteem as compared to normal children. They are also found to be socially immature and interpersonally dry. Spinal dysraphia affected children with hydrocephalus, shown to have difficulties in academics (Prakash et al., 2018). They have also shown deficits in abstract reasoning, executive functioning and low focus. Children having a positive environment at home and support from parents have known to improve in the living quality by 25%.

Management of the children affected with spinal dysraphism needs multidisciplinary approaches. Complete clinical investigation is mandatory (Tuite et al., 2018). The standard treatment for any type of spinal dysraphism is surgery after delivery (Peralta et al., 2020). The surgery is needed to control further damage to nervous tissue and manage infection. Pediatric neurosurgeons operate to seal the opening. Spinal cord and nerve roots are assembled back into the spine and are covered with meninges. In case of hydrocephalus, a shunt is surgically installed to remove excessive cerebrospinal fluids from the brain (Gordon et al., 2019). Parental counseling should be done for long-term management.

## Aim

The aims and objectives of the present study is to evaluate:

- The clinical presentation of the patients with spinal dysraphism
- Other associated congenital anomalies
- The surgical management and outcomes of patients with spinal dysraphism

## Materials and Methods

A study on surgical treatment of spinal dysraphism was done over the period of 6 months from December 2019- May 2020 at GMC,

Jammu. In the study at hand; the clinical aspects, management and surgical outcomes in case of spinal dysraphism were characterized. A total of 30 patients of spinal dysraphism were operated with 70% males and 30% females. The age group selected for the study was between 0-3 years.

In the present study, the most common site for swelling was found to be in lumbosacral region, followed by lumbar region, then thoracic region and lastly thoracolumbar region. No patient with cervical lesion was observed. Mothers underwent antenatal ultrasound to detect the anomaly.

After the patient was admitted, the following procedure was followed:

- The exposed nerves and tissue through the defect from trauma and infection was achieved by intravenous anti-biotic administration and saline dressing.
- All the patients were treated in pediatric surgery operation theater
- The patient was positioned in prone manner and general anesthesia was administered
- Elliptical incision was given over the swelling
- Then arachnoid was lifted in a circumferential fashion giving attention to the vascular supply to the placode.
- Careful and precise separation of the neural tissue from cutaneous tissue was done.
- A thorough search for associated anomalies such as tethering, diastematomyelia, thick filum terminale, dermal sinus tract was carried out and was corrected.
- Re-tethering was prevented by taking precautions such as: pial closure to cover the raw dorsal surface of the cord and/ or duroplasty to enlarge the subarachnoid spaces to facilitate free CSF around repair surface.
- After freeing neural placode, it was inverted and sutured.
- Finally, dura was closed and covered with overlying skin.
- Patients with large defects underwent flap repair.
- Majority of patients were put on Tablet Diamox for conservative management.
- In case of hydrocephalus, simultaneous ventriculo-peritoneal (V-P) shunting was done.
- The patients were nursed in prone position post-operatively to prevent wound contamination.
- IV antibiotics were given for 3-5 days post-operatively.
- They were regularly monitored on the basis of general conditions and systematic examination of motor, sensory and sphincteric functions.
- On 7th day patients were discharged.
- The follow up was made weekly for first month, then monthly for consecutive three months and three monthly thereafter.

## Results

A total of 30 patients were selected in this study. The age ranged from 1-3 years.

**Table 1: Sex, Age distribution and site of lesion of 30 patients taken for the present study**

Gender	No. of Cases
Males	21
Females	9
Site of lesion	No. of Cases

cervical	0
Thoracic	04
Thoracolumbar	02
Lumbar	05
Lumbosacral	19
<b>Age group</b>	<b>No. of Cases</b>
Neonates (<1 month)	17
Infants (1month-1 year)	6
Pre-school children (1-3 years)	2
School going children (>3 years)	5

Table 1 shows that the incidence is high in males (number of cases= 21), 70% when compared to females (number of cases=9), 30%. Children below the age of 1 month were mostly affected (number of cases= 17). Most of the patients were neonates, about 57% followed by infants upto 20%, pre-school children about 6% and school going children around 17%. In the present study, lumbosacral region was the most common site of swelling as seen in 63% patients followed by lumbar region (17%), thoracic region (14%) and thoracolumbar region (6%). No patient with cervical lesion was reported.

**Table 2: Various presentations of the patients with spinal dysraphism at the hospital**

S.No.	Clinical features		No. of cases	% of cases	
1	Swelling of back		28	94	
2	CSF leaks		14	47	
3	In continence	Both bladder and bowel	9	30	
		bladder	1	3	
		bowel	0	0	
4	Upper limb weakness		0	0	
	Upper limb deformity		0	0	
4	Lower limb weakness (power <3/5)	Hip	Right	9	30
			Left	9	30
		Knee	Right	15	50
			Left	13	43
		Ankle	Right	15	50
			Left	13	43
5	Lower limb deformity (club foot)	Bilateral	1	3	
		Right-sided	1	3	
		Left- sided	0	0	
6	Spine deformity	Absent vertebrae	7	23	
		Hemi-vertebrae	2	6	
		Kyphosis	0	0	
		scoliosis	1	3	
7	Hydrocephalus	Mild	7	23	
		Moderate	13	44	
		Severe	3	10	
8	Other stigmata	hypertrichiosis	2	6	
		Sacral dimple	2	6	
		Hemangiomatous patch	1	3	
9	Other associated anomalies	Anorectal malformation	1	3	
		Posterior urethral valve	1	3	
		hydroureteronephrosis	4	14	
		Chest wall deformity	1	3	
		Single kidney	1	3	
		hypospadias	2	6	
		Inguinal hernia	unilateral	2	6
			bilateral	1	3
		Dermal sinus	3	10	

In the current study, 94% patients presented to hospital reported swelling in the back, 47% patients reported with CSF leaks. In the present study, 30% patients reported with bladder and bowel incontinence, 3% had bladder incontinence but no case of bowel incontinence was reported. No patient with upper limb weakness or deformity was observed. Lower limb weakness and deformities were observed and reported in table 2. Hydrocephalus was reported in 77% patients. A majority of the patients responded well to the treatment of Diamox (acetyl choline esterase inhibitor). Other stigmata of spinal dysraphism are reported in table 2.

**Table 3: Power in lower limbs in patients with spinal dysraphism**

Region	Grade	No of cases on right side	No of cases on Left side
Hip	0	0	0
	1/5	3	5
	2/5	1	1
	3/5	5	3
	4/5	2	1
	5/5	19	20
Knee	0	6	6
	1/5	2	3

	2/5	3	0
	3/5	4	4
	4/5	3	3
	5/5	12	14
<b>Ankle</b>	0	6	6
	1/5	5	3
	2/5	3	1
	3/5	1	3
	4/5	3	3
	5/5	12	14

In the present study, no patient with 0 power at hip in both the lower limbs was observed. Various findings with power in lower limbs in patients with spinal dysraphism are reported in Table 3.

**Table 4: Antenatal ultrasound done in mothers to detect anomalies**

Antenatal ultrasound done		No of patients	%
Not done		6	20
Done	Anomaly detected	18	60
	Not detected	6	20

In the present study 80% mothers underwent antenatal ultrasound, the anomalies were diagnosed in 60% mothers whereas, in 20% mothers, no anomalies were detected (Table 4).

**Table 5: Folic acid intake of mothers**

Folic acid intake	No of cases	%
taken	16	53
Not taken	14	47

In the present study, 53% mothers were reported to be taking folic acid antenatally whereas, 47% mothers were not taking folic acid (Table 5).

**Table 6: Status of other siblings**

Status	No of cases	%
Affected/ Deceased	3	10
Healthy/ normal	17	57
Not applicable	10	33

It was reported that 57% patients had normal/ healthy siblings, while 10% patients had affected or deceased siblings and remaining 33% were of the first birth order (Table 6).

**Table 7: Diagnosis**

Diagnosis		No of cases	%
Spina bifida aperta	Myelomeningocele	17	57
	Meningocele	2	7
	Myelocele	1	3
	Rachschisis	2	7
Spina bifida occulta	Lipomeningomyelocele	3	10
	Hemangiomas patch with tethered cord	1	3
	Tethered cord with diastematomyelia	1	3
Combined defects	Myelomeningocele with dorsal tract	3	10

In the present study, 57% patients were found to have myelomeningocele, 7% had meningocele, 3% had myelocele and 7% had rachschisis. It was reported that 10% patients had lipomeningomyelocele, 3% patients had hemangiomas patch with tethered cord and 3% patients had tethered cord with diastematomyelia (table 7). The most common type of spinal

dysraphism was spina bifida aperta as observed in the patients under study.

**Table 8: Surgeries performed**

Procedure		No of cases	%
Excision repair of defect		25	84
Laminectomy and detethering of cord		4	13
Laminectomy and dissection of fibrous spur		1	3
Additional procedure with primary surgery	V-P shunting	3	10
	Excision of dermal sinus tract	3	10
Additional procedure in the follow-up period	V-P shunting	1	3
	Laminectomy and detethering of cord	1	3

In the current study, 83% patients underwent excision- repair of defect, 13% patients underwent laminectomy and detethering of cord, 3% patients had undergone laminectomy and dissection of fibrous spur. 10% patients underwent ventriculo-peritoneal shunting along with excision repair of defect and 3% patients underwent excision of dermal sinus tract. In the follow up period 3% patients underwent laminectomy and detethering of cord (Table 8).

**Table 9: Post-operative complications**

Complications		No of cases	%	
Immediate (First 24 hrs)	Vomiting	0	0	
	Fever	2	6	
	Seizures	1	3	
Early (1-7 days)	Meningitis	2	6	
	Wound infection	Minor	4	14
		Major	3	10
	CSF leak	4	14	
	Shunt blockage	1	3	
Late (after 7 days)	Hydrocephalus	1	3	
	Tethered cord	1	3	

In Post-operative study, 6% patients developed fever, 3% patients developed seizures in immediate post-operative period. In early post-operative period 6% patients developed meningitis, 14% patients developed minor wound infection, whereas, 10% patients developed major wound infection, 14% patients were reported with CSF leakage and 3% patients developed shunt blockage. In the late post-operative period, 3% patients developed hydrocephalus and 3% reported with tethering of cord (Table 9).

**Table 10: Outcomes at 3 month post-surgery**

Outcome	No of cases	%
Good	20	67
Satisfactory	6	20
Poor	4	13

In the present study, out of 30 patients operated for spinal dysraphism, 67% patients showed good outcome, 20% showed satisfactory outcome and 13% patients showed poor outcome after 3 months of surgery. Mortality rate was recorded to be 13% (Table 10).

## Discussion

Spinal dysraphism is a congenital anomaly encompassing wide spectrum of neural tube defects. With prenatal screening, the

incidence is on a decline as the patients can be tested early and further management could be planned accordingly to reduce the subsequent neurological, urological and orthopedic disabilities. In the present study, out of 30 patients, 21 were males and 9 were females. The data showed a higher risk of incidence in male than female. Similar findings were reported by Sherif and Momen, 2017, in a study of sixty patients with open spinal dysraphism, thirty-seven patients (61.7%) were males while twenty-three patients (38.3%) were females. Kumar and Singh 2003 reported that out of 155 patients of spinal dysraphism, the male to female ratio was 1.5:1. Spinal dysraphism was found to be more prevalent in infants (Choi et al., 2020). In the current study, the maximum number of patients belonged to neonatal age group. In a study done by Rao et al., 2015, the age of the patients ranges from one day to 17 years. The youngest patient was reported to be one day. In the present study, it was reported that the intake of folic acid by mothers reduced the risk of spinal dysraphism. Maternal preconception of folic acid has been found to reduce the risk of spina bifida (Dixon et al., 2019). In the present study, 10% patients had affected or deceased sibling. The risk of spinal dysraphism is greater if a woman has had previous fetal death (Guida et al., 2009). The commonest site of occurrence of spinal dysraphism was found to be in the lumbo sacral region (63%). This is in accordance with the study conducted by Premalal, 2018. He reported lumbosacral region as the most common site of occurrence of spinal dysraphism with 51.85%. In the present study, 30% patients showed weakness at hip, 50% patients and weakness in right knee and ankle and 43% patients reported weakness at left knee and ankle. Iftikhar and De Jesus, 2020, showed motor weakness and sensory deficit in the lower limbs of the patients. Sphinter dysfunction was seen in 30% patients in the present study. Sphinter dysfunction is reported to be a common occurrence in spinal dysraphism (Wide et al., 2020). In the present study, post-operative improvement was seen in 88% patients. In a study by Metcalfe et al., 2006, post-operative improvement in 72% patients was reported. In the current study, 77% patients showed hydrocephalus (Kafle et al., 2017). The most common associated anomaly with spinal dysraphism is hydrocephalus. Kumar and Singh, 2003 reported that hydrocephalus in 42.59% patients. Meningomyelocele was seen in 57% patients in the current study, followed by lipomeningomyelocele in 10% patients. The patients with lipomeningomyelocele were reported to have normal leg function and continence. The studies support direct injury to the protruding spinal cord as the primary cause of damage and loss of function. In the present study, 3 patients of lipomeningomyelocele were reported to have normal legs. Prenatal screening of fetus by ultrasound has lead to the detection of defect at an early stage in utero (Kumar et al., 2017). In current study, 80% mothers underwent ultrasound antenatally, out of which 60% were detected with anomalies. In the present study, 67% patients showed good outcome post operatively. A study by Borcek et al., 2012 reported 82.2% patients with good outcome post operatively. In the present study, we report a mortality rate of 13%.

## Conclusion

Spinal dysraphism is the most common type of congenital disability in children faced by pediatric neurosurgeon. Open type of spina bifida is seen to be more prevalent than closed type. From the current study it was concluded that spinal dysraphism is a complex disorder influencing multiple systems of body, thus, requires multidisciplinary approach for management and better outcome. However, surgery remains the treatment of choice along

with good post-operative care. Family support remains the most important aspect for the patient's better life.

## Ethical approval

The study was approved by the Institutional Ethics Committee.

## List of abbreviations

AFP: Alpha Feto Protein  
HCG: Human Chorionic Gonadotropin  
MRI: Magnetic Resonance Imaging  
CT: Computed Tomography  
CSF: Cerebrospinal Fluid

## Conflicts of interest

None declared

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## Authors' contributions

VKG and BG performed the operative interventions. AB and SS analyzed and interpreted the patients' data regarding the surgical management of spinal dysraphism. All the authors read and approved the final manuscript.

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