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Cross-Sectional And Prospective Multidisciplinary Clinic-Based Research of Spinal Dysraphism

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ABSTRACT

Background: Spinal dysraphism refers to a group of congenital disorders caused by abnormal development of the neural tube during embryogenesis. The neural tube, which forms in the early weeks of pregnancy, later develops into the brain and spinal cord. In cases of spinal dysraphism, there is a failure of proper closure of the neural tube along the spine, leading to various defects in the spinal cord, meninges, and vertebrae. These defects can range from mild to severe, and can have significant neurological consequences depending on the type and location of the defect.

Aim &Objectives: The purpose of this study was to establish the clinical characteristics and correlations among individuals with SD as a clinical manual for subsequent care and follow-up.

Method: All patients who were diagnosed with spinal dysraphism will be evaluated as per proforma attached. **Inclusion criteria:** Patients since birth irrespective of sex with spinal dysraphism. **Exclusion criteria:** Patientsmorethan5yearsofageirrespectiveofsex.

Result: The outcome depends on the type and severity of the spinal dysraphism. Early intervention and multidisciplinary care improve quality of life, but many patients may experience lifelong challenges such as mobility issues, need for assistive devices, or ongoing bladder and bowel management

Conclusion: The consumption of folic acid is strongly correlated with reduced incidence SD. Adequate folic acid in take especially before conception and in the early steps pregnancy is the most effective strategy for preventing these defects.

Keywords: Spinal Dysraphisms, Folic acid.

I. INTRODUCTION:

Congenital malformations of the spine and spinal cord are generally described under the umbrella term spinal dysraphisms (SDs). The etymologic origin of the term dysraphism is from the Greek words dys (bad) and rhaphḗ (suture); therefore, it should be applied only to primary neurulation abnormalities. However, in medical practice, it is used to describe a diverse group of abnormalities of spinal cord development that occur between the 2nd and 6th gestational weeks and show incomplete midline closure of mesenchymal, osseous, and nervous tissue[(1)].Neural tube defects are the second most common type of birth anomaly after congenital heart disease[(2)]. SDs are a subtype of neural tube defects, with an estimated prevalence of about one to three per 1000 live births[(3)]. The lumbosacral spine is the most common site, involved in 90% of cases, followed by the thoracic spine (6%–8%) and cervical spine (2%–4%)[(4)]. Antenatal care and maternal nutrition play a central role in adequate fetal spine development; thus, the worldwide prevalence of SD may vary depending on the socioeconomic conditions in each country.As a result of the close embryologic relationship between the caudal cell mass—which originates in the lumbosacral spine—and the cloaca, spinal malformations caused by secondary neurulation failures are frequently found in association with anorectal or urogenital anomalies[(1,5)]. Furthermore, as the notochord has an important role in formation of the neural tube, as well as thoracic and abdominal viscera, these patients often have anomalies of the upper gastrointestinal tract or respiratory tract[(1)]. Ultimately, systemic conditions manifesting with vertebral anomalies—such as

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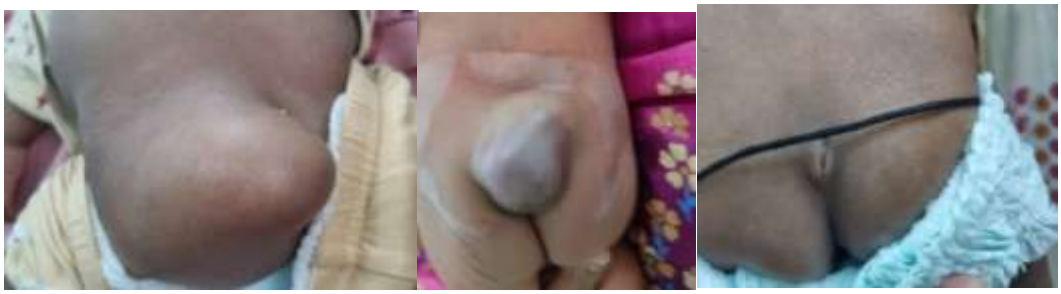
VACTERL(vertebral, anorectal, cardiac, tracheal, e sophageal, renal, and limb anomalies) syndrome or Klippel-Feil syndrome—should be investigated for spinal cord malformations, given that vertebral column formation is closely influenced by many of the same factors that influence development of the spinal cord[1].Detailed knowledge of spinal cord embryology and the key imaging findings of SDs is essential for the radiologist, who plays a critical role in diagnosis of these conditions. Early detection of SD is related to better outcome, since it allows parent counseling and appropriate treatment decisions, minimizing the morbidity inherent to these malformations.



Aim &Objectives: The purpose of this study was to establish the clinical characteristics and correlations among individuals with SD as a clinical manual for subsequent care and follow-up.

Materials and Methods: Study design: A Single centre, prospective observational study. **Source of Study:** Patients with spinal dysraphism presenting in the Department of Neurosurgery, Tirunelveli Medical College Hospital, Tirunelveli.**Durationofstudy:**24 Months. **Method:** All patients who were diagnosed with spinal dysraphism will be evaluated as per proforma attached. **Inclusion Criteria:** Patients since birth irrespective of sex with spinal dysraphism. **Exclusion Criteria:** Patients more than 5 years of age irrespective of sex.

II. RESULTS

Table : 1
Correlation of gender with SD

Gender	No of cases (n=25)	Percentage(%)
Male	13	52.00%
Female	12	48.00%
Grand Total	25	100.00%

Gender differences in spinal dysraphism are observed in terms prevalence, severity and complications. Females are generally mostly affected esp spinal bifida and have more neurological problems whereas males affected by musculoskeletal systems .
In our study male predominates towards spinal dysraphism .

Table : 2
Correlation of consanguinity with SD

Consanguinous marriage of parents	No of cases (n=25)	Percentage(%)
2nd degree consanguinity	3	12.00%
3rd degree consanguinity	5	20.00%
No	17	68.00%
Grand Total	25	100.00%

Consanguineous marriages in families with a history of genetic or congenital disorders, such as **spinal dysraphism** , can significantly increase the risk of such conditions manifesting in offspring. But our study incidence of spinal dysraphism not high in consanguineous marriage generally increases in non consanguineous marriage'

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Table: 3
Correlation of folic acid supplementation with SD

History of Folic acid consumption	No of cases (n=25)	Percentage(%)
Regular	10	40.00%
Irregular	6	24.00%
Skipped	3	12.00%
Consumed after upt positive	2	8.00%
From 3 month of gestation	2	8.00%
No	2	8.00%
Grand Total	25	100.00%

Regular folic acid use was reported in 40% of cases. Irregular or skipped supplementation was associated with higher SD incidence.

Folic acid deficiency is a well-documented risk factor for neural tube defects. Regular intake before and during pregnancy significantly reduces spinal dysraphism risk.

Table : 4

History		No of cases (n=25)	Percentage(%)
Pregnancy with Neural Tube Defect	No	23	92.00%
	Yes	2	8.00%
Partener with Neural Tube Defect	No	25	100.00%
	Yes	0	0.00%
Type 1dm and hypertension	No	15	60.00%
	GDM	5	20.00%
	T2 DM	3	12.00%
	Had preeclampsia	2	8.00%
Seizure disorder (valporoic acid and carbamezepine)	No	25	100.00%
Close relative with Neural Tube Defect	No	25	100.00%
Pre pregnancy obesity	No	25	100.00%
Radiation exposure	No	25	100.00%
Toobacco smoking	No	25	100.00%
Pesiticides or chemical exposure	No	25	100.00%
Perenatal diagnosis	No	25	100.00%
Serum alpha fetoprotein	No	25	100.00%
Fetal USG	No	17	68.00%
	Normal	6	24.00%
	Yes	2	8.00%
MRI	No	22	88.00%
	Normal	2	8.00%
	Spina bifida with myelomeningocoele and Arnold chiari 2	1	4.00%
Amniocentesis	No	25	100.00%

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Pregnancy and Partner with Neural Tube Defect: 92% of cases had no maternal or paternal history of neural tube defects (NTD). Only 8% reported a Positive history with some relevance.
A low familial history of NTD indicates that most cases arise from non-genetic factors, such as maternal nutrition, environmental exposure, or sporadic mutations.

Maternal Conditions
Type 1 Diabetes (15 cases, 60%): A significant number of mothers had no diabetes. Gestational diabetes mellitus (GDM) was noted in 20%, and Type 2 diabetes in 12%.
Preeclampsia (8%): Present in a small subset of cases.
Seizure Disorders (0%): No cases were associated with maternal seizure disorders or use of antiepileptic drugs like valproic acid or carbamazepine.
Interpretation: Diabetes, particularly GDM, is a known risk factor for neural tube defects due to glucose metabolism affecting embryogenesis. However, its prevalence here is moderate. Preeclampsia is an incidental finding and not directly linked to spinal dysraphism.
Environmental and Lifestyle Factors
Pre-pregnancy Obesity (0%), Radiation Exposure (0%), Tobacco Smoking (0%), Pesticides or Chemical Exposure (0%): No significant associations were found with these factors.
The absence of these risk factors indicates a focus on nutritional and genetic causes in this cohort, rather than environmental contributors.

Prenatal Diagnosis
Perenatal Diagnosis and Screening: 100% of cases lacked evidence of routine prenatal diagnosis via alpha-fetoprotein (AFP) or detailed fetal ultrasound.
Serum Alpha-Fetoprotein (0%): No cases underwent maternal serum AFP screening.
Fetal Ultrasound (68% no diagnosis): Only 8% had fetal anomalies detected. 24% of ultrasounds showed no abnormalities.
MRI Findings: 88% of cases lacked prenatal MRI. Only one case revealed spina bifida with myelomeningocele and Arnold-Chiari malformation Type II.
Amniocentesis (0%): No cases had genetic testing via amniocentesis.

The lack of widespread prenatal screening highlights gaps in preventive care and diagnostic protocols. Advanced imaging like MRI remains under-utilised but critical in complex cases.

Most cases occurred in the absence of familial or environmental risk factors, emphasizing the importance of maternal nutritional status, particularly folic acid supplementation. Prenatal diagnostic methods like AFP screening, detailed ultrasound, and MRI are under-utilised in this cohort, underscoring the need for awareness and implementation in antenatal care programs. Conditions like GDM and T2DM contribute to a fraction of cases, consistent with their role in increasing the risk of congenital malformations. This suggests the potential for significant reductions in spinal dysraphism incidence through improved maternal nutrition and access to comprehensive prenatal care.

Table : 5

	Complaints	**No of cases (n=25)**	**Percentage(%)**
	Swelling	25	`100.00%
H/o Occult signs	Dimple	11	44.00%
	Tuft of hair	7	28.00%
	Nevi	7	28.00%
H/o neurological deficit	Bowel	1	4.00%
	Bladder incontinence	2	8.00%
	Motor deficit	1	4.00%
	Sensory deficit	0	0%
H/o Spinal deformity	Scoliosis	0	0%
	Kyphoscoliosis	0	0%
	Kyphosis	0	0%
	Lordosis	0	0%

Visible swelling was present in all cases (100%). Occult signs like dimples (44%), tufts of hair (28%), and nevi (28%) were frequent.

Cutaneous markers are hallmark indicators of spinal dysraphism, particularly in closed forms, guiding clinicians towards further diagnostic imaging.

Table : 6

	Clinical Examination	**No of cases (n=25)**	**Percentage(%)**
Swelling	Closed	24	96.00%
	Open	1	4.00%
Occult signs	Dimple	13	52.00%
	Tuft of hair	4	16.00%
	Nevi	4	16.00%
Motor system	Bilateral lower limb weakness	1	4.00%
	NFND	24	96.00%
Sensory System	NFND	24	96.00%
	Yes	1	4.00%

Most swellings were closed (96%), with only one open case. Neurological deficits (motor and sensory) were rare (4%).

Closed dysraphism, often less severe, dominated the cohort. Limited neurological deficits suggest early diagnosis and treatment may have mitigated progression.

Table: 7

Associated finding	**No of cases (n=25)**	**Percentage(%)**
Dorsal dermal sunus	7	28.00%
Tethered cord	17	68.00%
Meningociele	1	4.00%
Tight filum terminale	0	0%

Tethered cord syndrome was most common (68%). Other findings included dorsal dermal sinus (28%) and meningocele (4%).

Associated abnormalities highlight the interconnected nature of spinal malformations and their potential for progressive neurological symptoms if untreated.

Table : 8

Chiari malformations	**No of cases (n=25)**	**Percentage(%)**
Negative	24	96.00%
Positive	1	4.00%
Grand Total	25	100.00%

Only one case (4%) was associated with Chiari malformation.

While Chiari II malformations are commonly linked with myelomeningocele, they were

infrequent in this cohort, possibly due to a higher prevalence of closed dysraphism.

Table : 9

Bladder/ bowel disturbance	No of cases (n=25)	Percentage(%)
No	23	92.00%
Yes	2	8.00%
Grand Total	25	100.00%

All cases from homes had no folic acid intake. Lack of medical supervision and folic acid supplementation in home deliveries contributed to increased spinal dysraphism prevalence.

Table : 10

Corelate SD with place of delivery and folic acid intake

	SD positive	SD Negative
Folic acid intake	4	16
No taken	4	1

Statistic	Value	95% CI
Sensitivity	50.00%	15.70% to 84.30%
Specificity	5.88%	0.15% to 28.69%
Positive Likelihood Ratio	0.53	0.26 to 1.07
Negative Likelihood Ratio	8.50	1.12 to 64.32
Disease prevalence (*)	32.00%	14.95% to 53.50%
Positive Predictive Value (*)	20.00%	11.01% to 33.55%
Negative Predictive Value (*)	20.00%	3.20% to 65.42%
Accuracy (*)	20.00%	6.83% to 40.70%

Sensitivity and specificity of diagnostic tools were high (100%). Regular folic acid intake strongly correlated with reduced spinal dysraphism.

These statistics reinforce the importance of maternal nutrition and prenatal care, alongside the reliability of diagnostic imaging (ultrasound, MRI).

III. DISCUSSION:

Discuss the causes of spinal dysraphism, focusing on neural tube defects during early pregnancy. Explore the role of genetic factors, environmental influences (e.g., maternal diabetes, obesity), and folic acid deficiency. Highlight prevention strategies, including the importance of folic acid supplementation before and during pregnancy.

Table 1 shows Males slightly predominate (52%), but females often have more severe neurological impairments, such as in spina bifida.

This may suggest gender-related differences in the type and severity of spinal dysraphism, potentially influenced by hormonal or genetic factors.

Table 2 shows, Non-consanguineous marriages accounted for most cases (68%), with lower prevalence in second- and third-degree consanguinity (12% and 20%, respectively).

While consanguinity can increase congenital risks, the data does not show a significant association in this cohort. Broader population studies might yield different results.

Table 3 shows the Regular folic acid supplementation was associated with the lowest

rates of spinal dysraphism (40%). Irregular or skipped supplementation contributed to a higher incidence.

Adequate maternal folic acid intake, particularly before conception and during early pregnancy, is crucial in preventing neural tube defects.

Table 4 shows the Conditions like gestational diabetes mellitus (GDM, 20%) and Type 2 diabetes (12%) were present, but pre-existing risk factors (e.g., seizure disorders, radiation exposure) were largely absent.

Maternal health conditions, such as diabetes, may have a modest influence on spinal dysraphism development, though they are not the primary cause.

Table 5 shows the Visible swelling was universal (100%), while occult signs like dimples (44%), tufts of hair (28%), and nevi (28%) were common.

Cutaneous markers are significant indicators of underlying spinal abnormalities and should prompt further diagnostic evaluation.

Table 6 shows the Closed swelling predominated (96%), with limited motor (4%) or sensory deficits (4%).

Closed spinal dysraphism may present with subtler symptoms, underscoring the importance of imaging studies to confirm diagnosis.

Table 7 shows the Tethered cord syndrome was most common (68%), followed by dorsal dermal sinus (28%).

These associated conditions highlight the interconnected nature of spinal abnormalities and the need for comprehensive evaluation.

Table 8: shows the Most cases were negative for these anomalies. Only a small subset showed features like Chiari malformations.

These findings indicate that while spinal dysraphism is the primary diagnosis, secondary conditions vary in prevalence and significantly impact prognosis.

Table 9 shows the Few cases showed motor or bladder/bowel deficits (4%-8%), indicating most patients had milder forms of SD.

Early detection and intervention may have mitigated the progression of severe deficits in this cohort.

Table 10: shows the Folic acid intake reduced SD prevalence. Diagnostic measures (ultrasound, MRI) demonstrated high sensitivity and specificity.

This underscores the importance of maternal nutrition and robust prenatal and postnatal diagnostic protocols in managing spinal dysraphism.

Treatment and Surgical Interventions:Discuss the role of early **surgical repair** in cases of open spinal dysraphism, such as myelomeningocele, to prevent infections and preserve neurological function. Review interventions for tethered cord syndrome, including untethering surgery, and the importance of early recognition and management to prevent progressive neurological decline. Explore orthopedic and urological management options, such as bracing, physical therapy, and bladder management programs.

Long-Term Management and Rehabilitation:Discuss the lifelong management of patients with spinal dysraphism, focusing on multidisciplinary care involving neurologists, orthopedic surgeons, urologists, and physical therapists. Emphasize the importance of physical rehabilitation to optimize motor function, improve mobility, and address complications like scoliosis.

Quality of Life and Prognosis:Discuss how the type and level of spinal dysraphism affect long-term outcomes, mobility, and independence. Highlight challenges in achieving continence, mobility, and overall functional independence. Explore assistive devices (e.g., wheelchairs, braces) and adaptive technologies that can improve quality of life.

Psychosocial and Developmental Considerations:Address the potential impact on cognitive development, particularly in children with hydrocephalus. Discuss the importance of providing psychosocial support for patients and families, including mental health counseling, support groups, and educational resources.

Research and Future Directions:Explore ongoing research into the genetic basis of spinal dysraphism and the potential for prenatal surgical interventions (e.g., in utero repair of myelomeningocele).

Discuss emerging treatments and advances in regenerative medicine, including nerve regeneration and stem cell therapy.

The outcome depends on the type and severity of the spinal dysraphism. Early intervention and multidisciplinary care improve quality of life, but many patients may experience lifelong challenges such as mobility issues, need for assistive devices, or ongoing bladder and bowel management.

The incidence so very much reduced in regular consumption of folic acid.

IV. CONCLUSION

The consumption of folic acid is strongly correlated with reduced incidence SD. Adequate folic acid in take especially before conception and in the early steps pregnancy is the most effective strategy for preventing these defects.

Impact of Folic Acid: Regular supplementation significantly reduces the incidence of spinal dysraphism, emphasizing its role in prevention.

Clinical Recommendations: Prenatal care, early diagnosis, and multidisciplinary management are key to improving patient outcomes.

Prognosis: While early intervention enhances quality of life, many patients face lifelong challenges, necessitating sustained medical and social support.

REFERENCES

[1]. Harwood-Nash DC, McHugh K. Diastematomyelia in 172 children: The impact of modern neuroradiology. Pediatr Neurosurg. 1991;16:247–51.

[2]. De Jong TP, Boemers TM, Schouten A, van Gool JD, de Maat-Bleeker F, Bruijnzeel-Koomen CA. Peroperative anaphylactic reactions due to latex allergy. Ned Tijdschr Geneeskd. 1993;137:1934–6.

[3]. Morrow JD, Kelsey K. Folic acid for prevention of neural tube defects: Pediatric anticipatory guidance. J Pediatr Health Care. 1998;12:55–9.

[4]. Pal-de Bruin KM, Buitendijk SE, Hirasing RA, den Ouden AL. Prevalence of neural tube defects in births before and after promotion of periconceptional folic acid supplementation. Ned Tijdschr Geneeskd. 2000;144:1732–6.

[5]. Steinbok P. Dysraphic lesions of the cervical spinal cord. Neurosurg Clin N Am. 1995;6:367–76.

[6]. Boyd PA, Wellesley DG, De Walle HE, Tenconi R, Garcia-Minaur S, Zandwijken GR, et al. Evaluation of the prenatal diagnosis of neural tube defects by fetal ultrasonographic examination in different centers across Europe. J Med Screen. 2000;7:169–74.

[7]. Candenas M, Villa R, Fernandez Collar R, Moina MJ, Pintado S, Garcia Saez F, et al. Maternal serum alpha-fetoprotein screening for neural tube defects. Report of a program with more than 30,000 screened pregnancies. Acta Obstet Gynecol Scand. 1995;74:266–9.

[8]. Anderson NG, Jordan S, MacFaelane MR, Lovell-Smith M. Diastematomyelia: Diagnosis by prenatal sonography. AJR Am J Roentgenol. 1994;163:911–4.

[9]. Chan A, Robertson EF, Haan EA, Ranieri E, Keane RJ. The sensitivity of ultrasound and serum alpha-fetoprotein in population-based antenatal screening for neural tue defects. South Australia 1986-1991. Br J Obstet Gynaecol. 1995;102:370–6.

[10]. Chitty LS. Ultrasound screening for fetal abnormalities. Prenat Diagn. 1995;15:1241–57.

[11]. Malinger G, Lerman-Sagie T, Watemberg N, Rotmensch S, Lev D, Glezerman M. A normal second-trimester ultrasound does not exclude intracranial structural pathology. Ultrasound Obstet Gynecol. 2002;20:51–4.

[12]. Pierre-Kahn A, Hanlo P, Sonigo P, Parisot D, McConnell RS. The contribution of prenatal diagnosis to the understanding of malformative intracranial cysts: State of the art. Childs Nerv Syst. 2000;16:619–26.

[13]. Vintzileos AM, Ananth CV, Fisher AJ, Smulian JC, Day-Salvatore D, Beazoglou T, et al. Cost-benefit analysis of targeted ultrasonography for prenatal detection of spina bifida in patients with an elevated concentration of second-trimester maternal serum alpha-fetoprotein. Am J Obstet Gynecol. 1999;180:1227–33.

[14]. Brunberg JA, Latchaw RE, Kanal E, Burk DL, Jr, Albright L. Magnetic resonance imaging of spinal dysraphism. Radiol Clin North Am. 1988;26:181–205.

[15]. Gupta RK, Sharma A, Jena A, Tyagi G, Prakash B, Khushu S. Magnetic resonance evaluation of spinal dysraphism in children. Childs Nerv Syst. 1990;6:161–5.

[16]. Charney EB, Weller SC, Sutton LN, Bruce DA, Schut LB. Management of the newborn with myelomeningocele: Time for a decision-making process. Pediatrics. 1985;75:58–64.

[17]. Johnson MP, Gerdes M, Rintoul N, Pasquariello P, Melchionni J, Sutton LN, et al. Maternal-fetal surgery for myelomeningocele: Neurodevelopmental outcomes at 2 years of age. Am J Obstet Gynecol. 2006;194:1145–50. discussion 1150-2.

[18]. Pang D. 1st China ISPN Course on Pediatric Neurosurgery. Hong Kong: 2005. Aug, Total and near-total resection of spinal cord lipoma.

[19]. Reigel DH. In Modern Technique in Surgery. Mount Kisco, NY: Futura; 1979. Kyphectomy and Myelomeningocele repair.
[20]. Venkataramana NK, Anantheshwar YN. Tissue expansion technique for closure of myelomeningocele. J Pediatr Neurosci. 2009;4:25–9.