"ROLE OF MRI IN CHARACTERIZATION AND CATEGORIZATION OF THE SITE AND TYPE OF SPINAL DYSRAPHISM – A ONE YEAR HOSPITAL BASED OBSERVATIONAL STUDY"

BY

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Sub: Institutional Ethical Clearance for the study.

With reference to the above, we wish to inform you that your proposed research project titled **"ROLE OF MRI IN CHARACTERIZATION AND CATEGORIZED OF THE SITE AND TYPE OF SPINAL DYSRAPHISM- A ONE YEAR HOSPITAL BASED OBSERVATIONAL STUDY"** is ethical and justifiable. The proposed research project has been cleared by the JNMC Institutional Ethics Committee.

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ABSTRACT

BACKGROUND

Congenital conditions that cause aberrant growth in the spine and/or spinal cord are referred to as spinal dysraphism. Ectodermal, mesodermal, and neuroectodermal tissue maldevelopment is the root cause of it.

To diagnose spinal dysraphism, a number of diagnostic modalities are available, including amniocentesis, USG, CT, and MRI. Among them, intracranial fluid-containing areas, including the ventricles, arachnoid cysts, and extra-axial fluid collections, can be measured and tracked in size using magnetic resonance imaging (MRI). MRI has helped with early diagnosis and characterization of SD for deciding early and individually tailored treatment plan.

OBJECTIVE

The objective of this study is to characterize and categorize the site and type of spinal dysraphism as seen on magnetic resonance imaging (MRI). We also assessed for the additional/ associated findings in cases of spinal dysraphism.

MATERIALS AND METHODS

We conducted a Prospective one year hospital based observational study by including 55 patients referred with clinical suspicion or previously diagnosed of spinal dysraphism referred for MRI SPINE to the department of Radio-diagnosis at KLE'S Prabhakar Kore Hospital and Medical Research Centre, Belagavi. Universal sampling method was used. We characterized and classified the spinal dysraphisms according to the new anatomical-clinicoradiological classification.

RESULTS

We had aimed at analysing the distribution pattern of SD changes on MRI among the suspected cases of SD. We had included 55 patients irrespective of the age and gender. Those aged between 1.1 to 12 months were more with incidence of 30.9% (17/55) followed by 20% (11/55) each aged </=1month and > 5 years. 6 (10.9%), 4 (7.3%), 3 (5.5%) and 2 (3.6%) were aged between 36.1 to 48 months, 12.1 to 24 months, 48.1 to 60 months and 24.1 to 36 months respectively. There was statistically significant association with age or gender distribution observed.

CONCLUSION

In a study population of 55, the average age of our patients who presented with spinal dysraphism was 29.91±8.6 months. Minimum age being 15 days and maximum was 96 months. 29 (52.7%) males and 26 (47.3%) females were observed. Maternal malnutrition was the commonest predisposing factor among 27.3% followed by Genetic predilection among 23.6%. Lower lumbar swelling among 40% (22 cases) was the most common symptom. Patients with multiple spinal and vertebral changes were more accounting for 40% of the cases. 18.88% had associated vertebral deformities. 5 (9.1%) were found with renal anomaly.

KEYWORDS: Spinal dysrapism, MRI, associated findings in SD

LIST OF ABBREVIATIONS

SD	SPINAL DYSRAPHISM
OSD	OCCULT SPINAL DYSRAPHISM
USG	ULTRASONOGRAPHY
CT	COMPUTED TOMOGRAPHY
MRI	MAGNETIC RESONANCE IMAGING
CNS	CENTRAL NERVOUS SYSTEM
PNS	PERIPHERAL NERVOUS SYSTEM
CSF	CEREBRAL SPINAL FLUID
MTHFR	METHYLENETETRAHYDROFOLATE REDUCTASE
AFP	ALPHA-FETOPROTEIN

TABLE OF CONTENTS

LIST OF IMAGES

LIST OF GRAPHS

LIST OF TABLES

INTRODUCTION

Congenital conditions that cause aberrant growth in the spine and/or spinal cord are referred to as spinal dysraphism. Ectodermal, mesodermal, and neuroectodermal tissue maldevelopment is the root cause of it.[1, 2] Based on the appearance, there are two main forms of spinal dysraphism: occulta (closed) if the spinal lesion is not evident on the surface and aperta (open) if it is. Meningocele, myelomeningocele, lipomeningocele, lipomyelomenigocele, myeloschisis, and rachischisis are common presentations.[3]

Three stages are involved in the development of the spinal cord [4]

(1) Gastrulation, which results in the formation of the notochord

(2) Primary neurilation, which produces the upper nine tenths of spinal cord; and

(3) Secondary neurilation & retrogressive differentiation, which leads to the development of the conus tip as well as filum terminale.

A range of congenital conditions resulting from aberrations in any of the previously mentioned phases are referred to as spinal dysraphism. Consequently, there is either no fusion or incomplete fusion of the midline spinal components.[4, 5]

Adequate folic acid consumption throughout pregnancy has been demonstrated to dramatically lower the incidence of spinal dysraphism. Racial backgrounds and genetic inheritance are other possible etiologies. Infants with spinal dysraphism tend to be born to mothers who have not experienced any spinal dysraphism in their previous offsprings.[5] Nevertheless, the chance of having further children with spinal dysraphism rises to 1 in 20 following the birth of the first offspring with spinal dysraphism. Certain newborns with

spinal dysraphism may present with trisomies, duplications, deletions, and single-gene mutations, among other chromosomal abnormalities.[6]

Pregestational diabetes may raise the probability of spinal dysraphism and other abnormalities of the central nervous system, according to few reports. Considerable morbidity as well as mortality are associated with this condition. About 0.05 to 0.25 per 1000 live births is the estimated incidence of spinal dysraphism. The prevalence has decreased over the past 25 years due to comprehensive prenatal screening and folic acid supplementation. [7]

Nutritional deficiency accounts for up to 50% of all cases of spinal dysraphism, making it the most often reported risk factor. Zinc deficiency has also been linked to this illness in addition to folic acid deficit. Spinal dysraphism can also result from consuming certain substances in excess. A lack or excess of vitamin A may contribute to the development of spinal dysraphism. Furthermore, nitrates are closely linked to risk factors and can be found in groundwater and canned meat. Ingesting the fungal metabolite cytochalasin can also result in spinal dysraphism. Additionally, a number of drugs have been linked to spinal dysraphism. For instance, when used during pregnancy, the well-known antiepileptic medication sodium valproate has been linked to a 1% to 2% risk of myelomeningocele formation.[8, 9]

There are 0.5 to 8 incidences of both open and closed spinal dysraphism for every 1,000 live births. There are notable regional differences. The incidence is higher in developing countries, as expected.[2,10] But with better nutrition and increased knowledge regarding folic acid intake both prior to and during pregnancy, the incidence has declined

over time. The lumbar & sacral regions of the spine are most frequently affected by this condition, with the cervical spine being least affected. [11]

The cervical spine is affected by 0–5%, the thoracic spine by 5%–10%, the lumbar spine by 20–30%, the lumbosacral spine by 30–50%, and the sacral spine by 5–15%. Uncommon in the cervicothoracic area. [12]

To diagnose spinal dysraphism, a number of diagnostic modalities are available, including amniocentesis, USG, CT, and MRI. Among them, intracranial fluid-containing areas, including the ventricles, arachnoid cysts, and extra-axial fluid collections, can be measured and tracked in size using magnetic resonance imaging (MRI). The majority of closed spinal dysraphisms are asymptomatic at birth and are only suspected when cutaneous indicators or clinical signs of neurological abnormalities appear later in life. In the meantime, MRI has helped with early diagnosis of these conditions and more individualized treatment plan.[13,14]

Meanwhile, MRI has also enabled in both judgment of these syndromes and earlier &individually personalized conduct. For the evaluation of spinal dysraphism, The spinal cord, subarachnoid space, vertebral bodies, and intervertebral discs can all be clearly defined with the use of MRI, which is safe to use on newborns and children without posing a risk of exposure to ionizing radiation.[13, 14] In order to analyze the incidence of SD in our demographic area, the current study was carried out.

OBJECTIVES

Primary objective- The objective of this study is to characterize and categorize the site and type of spinal dysraphism as seen on magnetic resonance imaging (MRI).

Secondary objective- To study the additional/ associated findings in cases of spinal dysraphism.

REVIEW OF LITERATURE

Headings

1) History of spinal dysraphism
2) Definition of spinal dysraphism
3) Epidemiology of spinal dysraphism
4) Anatomy and embryology of spinal cord
5) Clinical significance
6) Types of spinal dysraphism
7) Etiology of spinal dysraphism
8) Diagnostic tests for spinal dysraphism

HISTORY OF SPINAL DYSRAPHISM

Spina bifida was first discovered by Hippocrates,

- The first definitive description of spina bifida was made by the Dutch clinician Pieter van Foreest (1522–1597) in the late 1500s.
- In 1614: Nicolaas Tulp (Claes Piereszoon) --- first coined “spina bifida”. Ligating/ amputating the dural sac --- surgical management of spina bifida over centuries. The outcome of this technique constantly fatal as damage of central spinal cord and fluid leak or hydrocephalus.
- In 1918, scholar Charles H. Frazier: surgical mode for repairing spina bifida, with multilayer closures using fascia, muscles, dura, and skin advocated.

- In 1967: Sharrard, confirmed that improved outcomes by carrying out the surgery of spina bifida in the perinatal period. Now, the surgery of spina bifida in the perinatal period, is the preferred treatment.[15]

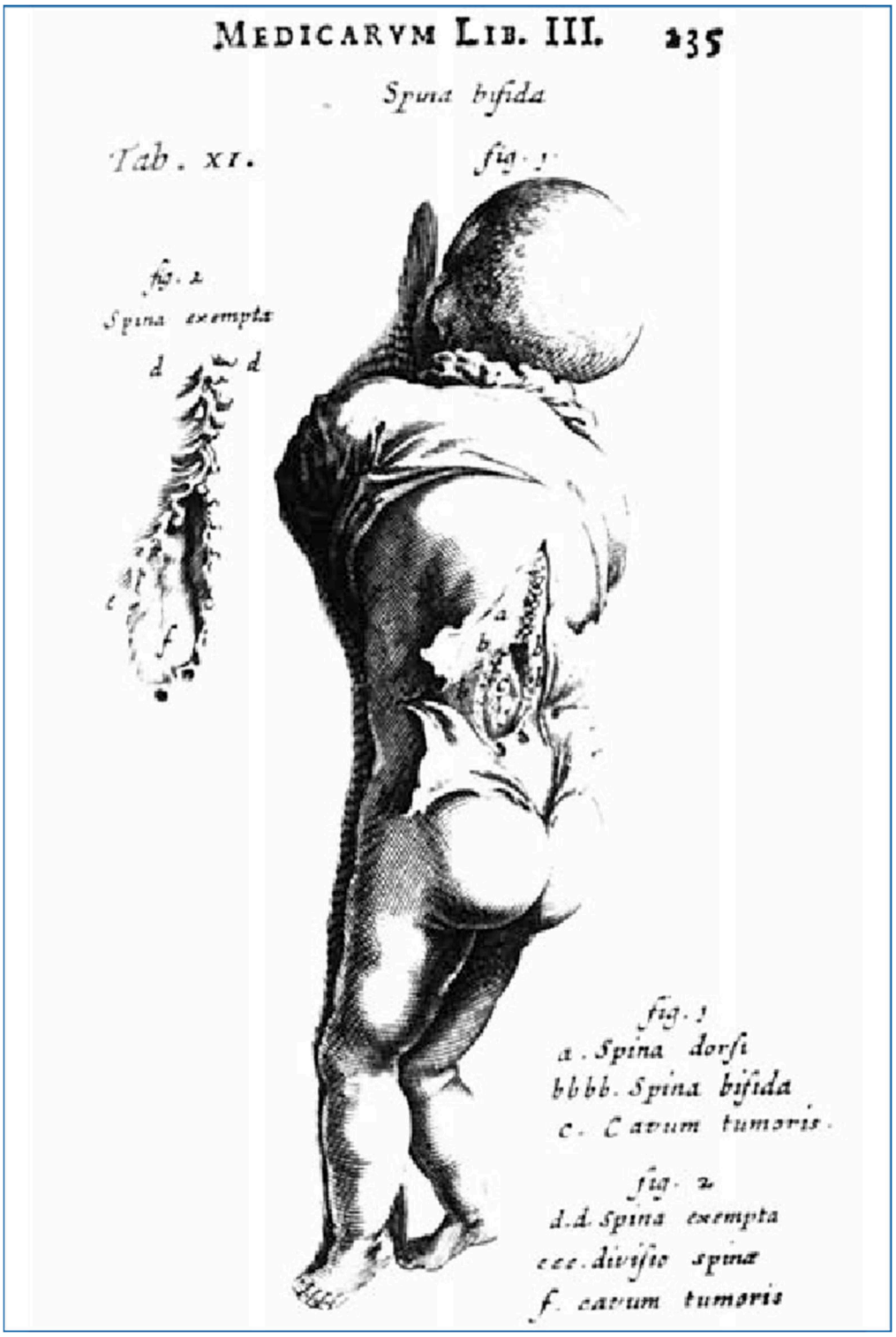


Figure 1: First ever case of SD reported

SPINAL DYSRAPHISM

A range of congenital defects known as spinal dysraphism cause a damaged neural arch, which allows meninges or other neural components to herniate and cause a range of clinical manifestations. They are divided into two groups: occulta, which present without an external lesion, and aperta, which has a visible lesion.[1-4]

EPIDEMIOLOGY

Spinal dysraphism is thought to affect 1-3 out of every 1000 live newborns. Over the past few decades, improvements in prenatal care, folic acid supplements, high-resolution ultrasonography for prenatal screening, and biochemical indicators have all contributed to a global drop in the prevalence of spinal dysraphism.[15]

In a retrospective examination of anatomical-clinic radiological analysis, Chellathurai A et al. found that a significant percentage of instances of spinal lipomas, including posterior myelomeningocele (14.2%) and lipomyeloceles and lipo-myelomeningoceles (31.3%), were reported. On the other hand, the least common cases were of anterior myelocoele (0.2%), sacral chordoma (0.2%), and intrasacral meningocele (0.2%). On the basis of the examination of the collected data, a new classification was suggested.[16]

After reviewing 247 cases, Johari AN et al. discovered that congenital kyphosis—18—and scoliosis—229—were the most common. Seven years was the average age. In the congenital scoliosis group, the mean Cobb angle at presentation was 42.45° (range 5 to 97°± 20.09) for those with normal MRIs, and 49.4° (range 8 to 145°± 23.77) for those with abnormal MRIs. The mean K angle at presentation for the congenital kyphosis group was 47.7° (range 14 to 110°, SD 33.33) for the individuals with abnormal MRI & 47.36°

(range 15 to 70°, SD 16.63) for the individuals with normal MRI. One-thirty patients had abnormal MRI results (congenital kyphosis, 38.8%, and congenital scoliosis, 53.7%). The segmentation (66.6%) and mixed (65%) kinds exhibited the highest frequency of aberrant MRI findings.

The dorsal region exhibited the highest incidence (61.9%) of deformities. The two most prevalent types of dysraphism were tethered cord and diastematomyelia. The presence of dysraphism and the type of deformity were significantly correlated.[17]

Agrawal A et al examined 28 children (17 boys and 11 girls) who underwent spinal dysraphism surgery during the study period. The age range for the median was 1 day to 6 years, or 120 days. Only thirteen moms received folic acid supplements during pregnancy, and fifteen mothers did not seek any routine prenatal care. Thirteen babies were born at a hospital, while fifteen babies were born at home. The lumbosacral area was the most frequent location (67.8%). At the time of presentation, seven patients had sac rupture, one kid had a localized infection, and four patients had hydrocephalus, which necessitated a shunt before surgical repair. At follow-up, two patients had hydrocephalus and required shunt surgery. The average duration of hospital stay was 7 days (interval: 5–31 days; median, 10 days).[18]

In a study of 6558 individuals, Choi SJ et al. found that 2.8% of cases with cutaneous stigmata had a pooled proportion of OSD, and 0.6% of patients had neurological surgery to treat their illness. Patients with both an unusual dimple and mixed stigmata showed a greater correlation with OSD.[19]

ANATOMY OF SPINAL CORD

The spinal cord is a well-organized, cylindrical structure. It starts at the base of the skull at the foramen magnum, where the medulla oblongata continues. It's situated inside the spinal canal, or vertebral column. It reaches up to 45 cm in men and up to 43 cm in women.

There are 31 segments that make up the spinal cord: cervical 8, thoracic 12, lumbar 5, sacral 5, and coccygeal 1. Thirty-one pairs of spinal nerves and their corresponding spinal root ganglia make up these segments. The motor, sensory, and autonomic fibers are found in spinal nerves. The intervertebral foramen is where these nerves exit.[20, 21]

For the brachial and lumbosacral plexus, the spinal cord has two major enlargements at the cervical and lumbar regions. Breadth of the spinal cord varies from 0.64 to 0.83 cm in the thoracic area and from 1.27 to 1.33 cm in the cervical and lumbar regions. With 13.3 +/- 2.2 mm, the segment at C5 has the biggest transverse diameter. At T8, it drops to 8.3 +/- 2.1 mm, and at L3, it rises to 9.4 +/- 1.5 mm. The anteroposterior diameter is less variable.[22]

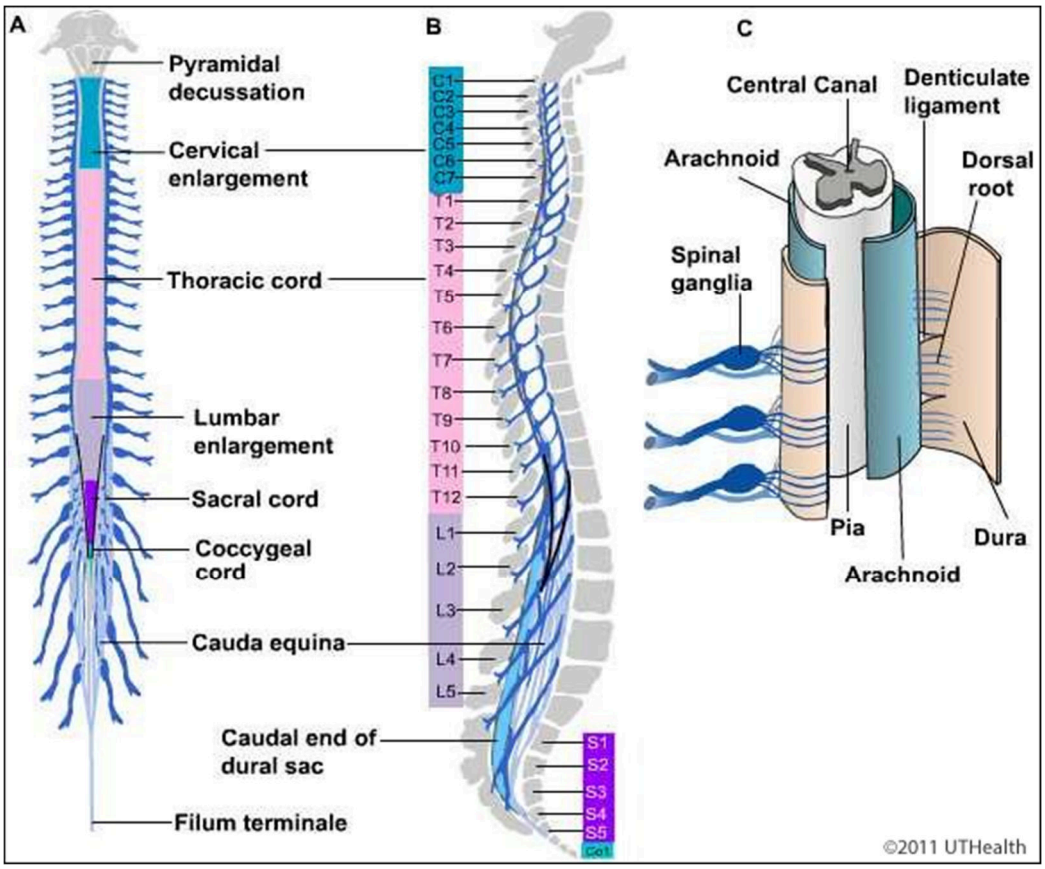


Figure 2: Schematic representation of the vertebral column[20]

VERTEBRAL CANAL

The vertebral foramen of cervical, thoracic, and lumbar vertebrae constitute the vertebral canal. The level of the L2 vertebra is usually where the spinal cord ends, giving rise to the cauda equina, a network of branching spinal nerves and nerve rootlets. The second through fifth lumbar vertebral levels are traversed by these spinal nerves.

The vertebral foramen formed by the posterior serving of the vertebral body, the pedicles, the bilateral laminae, and the assembly of the laminae at the transverse process. The only exemptions to this anatomical founding are the atlas and axis (C1 and C2 vertebrae) considered atypical of the cervical spine. These two vertebrae lack a vertebral body; the atlas contains an anterior and posterior arch, and the anatomy of the axis is made up of the odontoid process that projects superiorly into the vertebral foramen of the atlas.

In addition, the transverse processes of the cervical vertebrae contain an opening- the transverse foramina (plural of foramen).The transverse foramina of C1-C6 transmit the vertebral arteries. Those of C1-C7 transmit the vertebral veins. [20, 23]

The vertebral canal is surrounded by two essential structural ligaments. The posterior longitudinal ligament and the ligamentum flavum are two ligaments. Ligamentum flavum joins the laminae of each vertebra and runs the length of the spinal column. The anterior border of the vertebral canal is the posterior part of the vertebral body, and it is accompanied by the posterior longitudinal ligament.[23, 24]

The anterior longitudinal ligament links the bodies of the vertebrae and avoids anterior dislodgment of the vertebrae. It can be dented or torn in a stroke injury, violent---hyperextension of the neck, as recurrently seen in a collide vehicle accident, might damage the anterior longitudinal ligament and crush the posterior portions of cervical vertebrae.

The spinal canal diameter varies with each vertebral region, including the cervical, thoracic, and lumbar regions. The average diameter of the cervical vertebrae is 17 mm. The thoracic vertebral canal varies, with each vertebra moving in a cranial-caudal direction. Starting at the T1 vertebral level, the spinal canal diameter is 16 mm on average, and the diameter begins to decrease at the T2 level to 14 to 15 mm. The T4 vertebra has the smallest diameter moving caudally; the spinal canal diameter from T3-T11 remains relatively stable at 15 mm. At the T12 vertebra, the spinal canal diameter increases to an average of 18 mm. Compared to the cervical and thoracic regions, the lumbar vertebral group has a larger spinal canal diameter, on average, with the L5 diameter being the largest of the lumbar vertebrae at around 17.5 mm.[20-24]

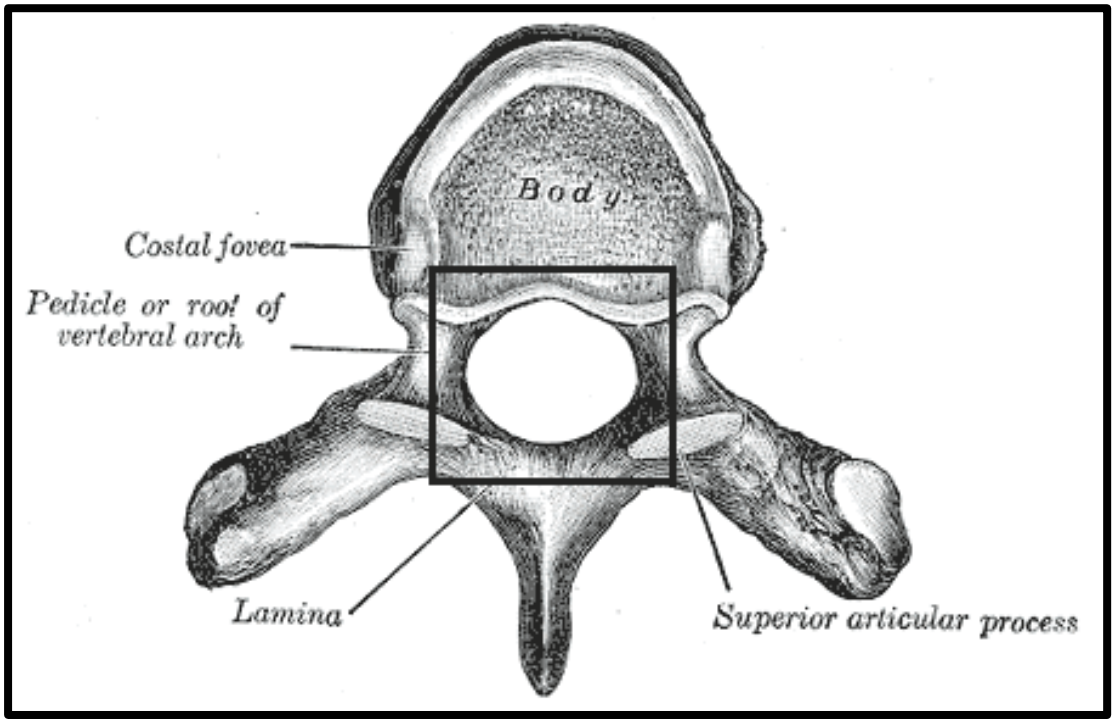


Figure 3: Schematic representation of vertebra[25]

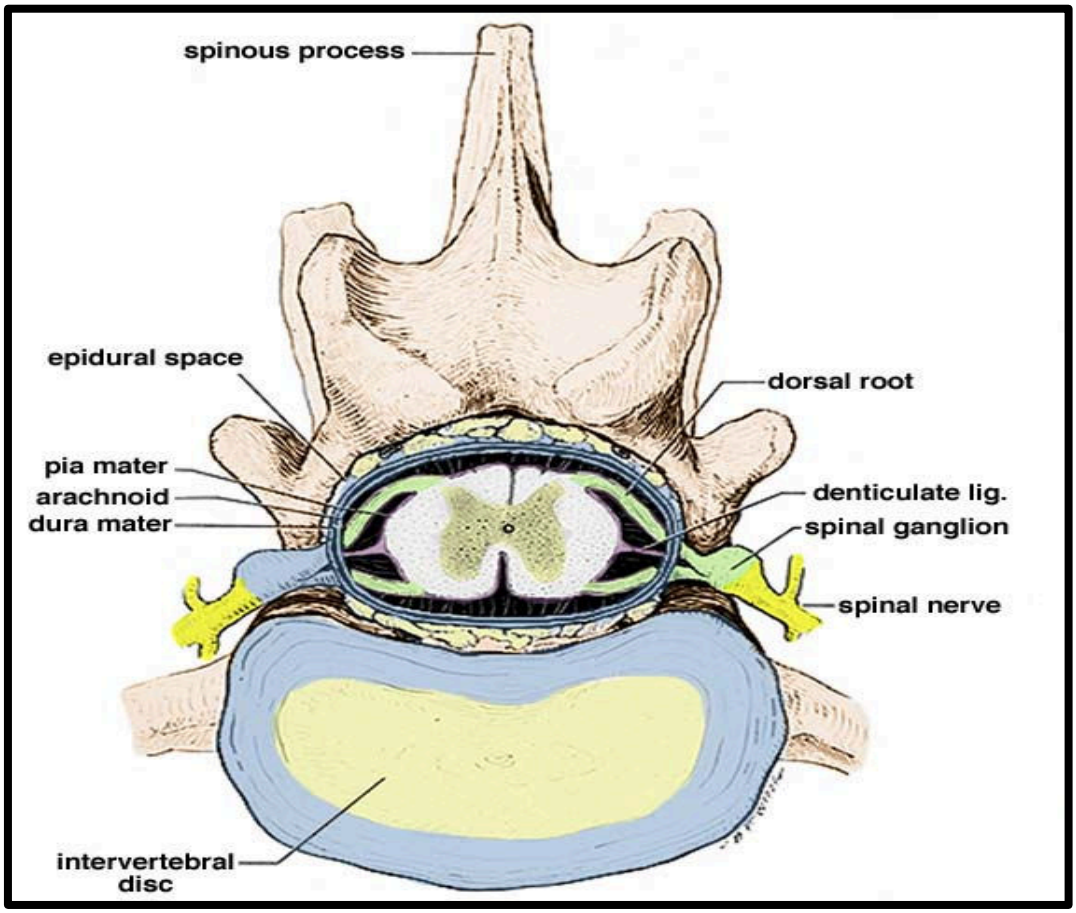


Figure 4: Parts of vertebra[26]

EMBRYOLOGY

The three germinal layers of the CNS system are the ectoderm, mesoderm, and endoderm.[27, 28, 29]

1. The primary initiator of the CNS's embryogenesis is the ectoderm. One particular subtype of the ectoderm is the (1) surface ectoderm, from which arises to the epidermis, hair and nails. In addition, ectoderm is subspecialized to develop into the (2) neural ectoderm, that develops into the neural tube aswell as the neural crest, which further develops and differentiates to form the brain, spinal cord, and peripheral nerves.

2. The lining of the respiratory and digestive systems originates from the endoderm. Additionally, it gives rise to the pancreas, bladder, and liver, among other abdominal organs.

3. There are three distinct parts to the mesoderm:

   ●*Paraxial mesoderm*: The axial skeleton, muscle aswell as the dermis originate from somites found in this mesoderm.

   ● *Intermediate mesoderm*: This mesoderm layer develops and differentiates to form the kidneys, urogenital structures, and gonads.

   ● *Lateral plate mesoderm*: is eventually divided into parietal & visceral mesoderm, which further forms the limb skeleton and muscular component of the intestinal tube respectively.

Spinal Cord

The spinal cord is formed from the neural plate, now contains 3 layers;[30]

1. *Ventricular layer* that lines the central canal
2. *Mantle layer* that contains neuronal bodies, which will eventually form the gray matter
3. *Marginal layer* that contains axons, and will eventually form the white matter

While this article summarizes the embryological changes that occur within the CNS, the peripheral nervous system (PNS) is formed from neuroepithelial cells. These cells travel from the pia mater to the ventricular layer of the spinal cord, where they differentiate and migrate to form glioblasts, neurons, and ependymal cells. As this information is often tested on boards, that myelin sheath, a sheath composed of support cells, wraps around axons and insulates neurons to increase the speed of neuronal conduction.

1. The *neurolemma* that originates from Schwann cells (that are derived off the neural crest cells), myelinates peripheral axons.
2. *Oligodendrocytes*, that are derived from neuroepithelium, myelinate CNS axons.

The entire CNS is covered in three membrane layers:

1. Dura mater: made of the surrounding mesenchyma, it is robust and sturdy.

2. Arachnoid mater: produced from the neural crest cells; combines with Pia mater to form a single layer.

3. Pia mater: closely covers the central nervous system; originates from the neural crest.

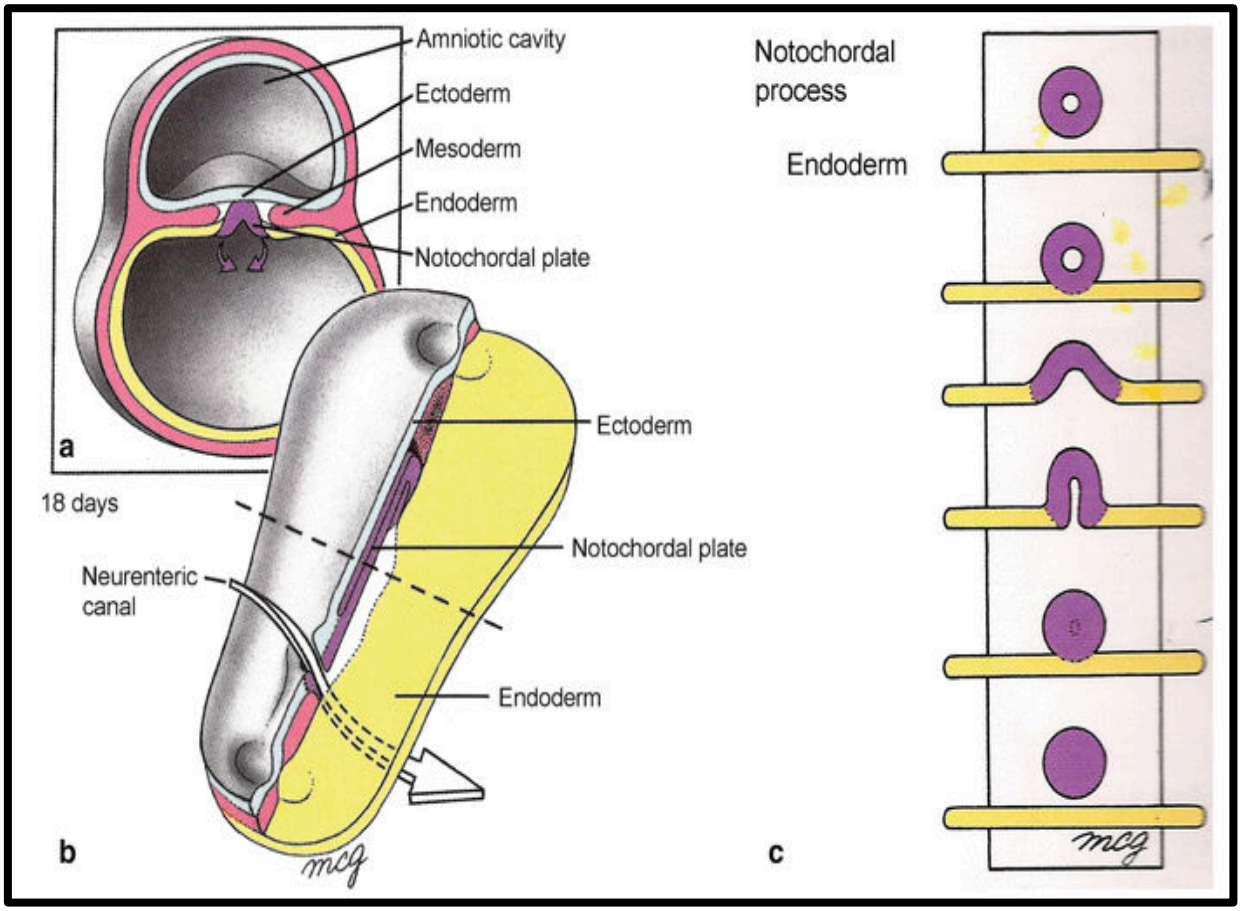


Figure 5: Embryology of CNS formation [31]

CLINICAL SIGNIFICANCE

Complexity in the process of embryogenesis can lead to subtle or severe abnormalities (pathophysiological alterations).

- ***Teratogenesis*** is the term used to describe any external stimulus that has the potential to affect an embryo's growth. Between weeks 3 and 8, embryos are extremely vulnerable as it is the crucial time of organogenesis.[32]
- The inability of symmetrical halves of an anatomical structure to fuse together is known as **dysraphism**. Spina bifida deformities are among them, although they're not the only ones.[33]

1. When the vertebral column fails to fuse but the other layers continue to develop normally, *spina bifida occulta* ensues. This type of dysraphism is considered the least severe and typically affects the lumbosacral area, specifically the S1 and S2 levels. In the field of abnormality, it can be linked to moles, angiomas, lipomas, and abnormal development of hair.

2. When there is an inadequate fusion of skin, with or without a cyst, *spina bifida aperta* might occur. Because the arachnoid membrane still covers the spinal cord, subarachnoid space is preserved and CSF leaking is prevented.

3. Among dysraphisms, *spina bifida cystica* is the most severe. Urinary or fecal incontinence may develop in these patients. Eighty percent of these lesions develop in the lumbosacral area.[33]

Dysraphism occurring in the cranium causes malformations similar to spina bifida:

1. The brain parenchyma extends into the subarachnoid space in an encephalocele. The Chiari III deformity, which results in the spinal cord twisting and a portion of the cerebellum protruding, may be connected to it. Cleft lip and palate are commonly associated with this. [34]
2. 2. Anencephaly: The cerebral cortex and thalamic structures are typically absent, but the cerebellum, brainstem, and spinal cord are present (albeit perhaps abnormal). The explanation could be the failure of notochord signaling, which is necessary for the activation of neural crest cell maturation or the establishment of the median hinge point..[35]
3. *Holoprosencephaly:* Failed features of midline structure. Features include a single central incisor, cyclopia, or unpaired cerebral hemisphere.[36]

4. *Craniorachischisis totalis* is when the entire neural plate fails to fold, and CNS is open to the amniotic cavity. These are often associated with still-born fetuses.[37]

TYPES OF SPINAL DYSRAPHISM [6, 38-40]

Open types

- Cranial
- Anencephaly
- Spinal (myelomeningocele)
- Aperta
- Cystica
- Craniorachischisis

Closed types:

- Posterior vertebral body fusion defect without spinal cord malformation (so-called *spina bifida occulta*)
- Forms of occult spinal dysraphism
- Frequently with overlying cutaneous and/or subcutaneous abnormalities
- Lipomyelomeningocele and lipomeningocele
- Diastematomyelia and other split cord malformations
- Dermal sinus tracts
- Often without overlying cutaneous and/or subcutaneous abnormalities
- Spinal epidermoids and dermoids
- Thickened and/or fatty filum terminale

The new anatomical-clinicoradiological classification of spinal dysraphism is as follows:[16]

1) **POSTERIOR SPINAL DYSRAPHISM**

Posterior: Open spinal dysraphism:

- Myelomeningocele
- Myelocele
- Hemi myelomeningocele
- Hemi myelocele

Posterior: Closed spinal dysraphism

- **With subcutaneous mass:**

- Meningocele
- Saccular limited dorsal myeloschisis
- Myelocystocele
- Posterior lipomyelocele (spinal lipoma Types IB, IIB, IIIB,)
- Posterior lipomyelomeningocele (spinal lipoma type VB)

- **Without subcutaneous mass:**

With cutaneous stigmata

- Other than a dermal sinus: Non-saccular limited dorsal
- Myeloschisis
- Post vertebral neurenteric cysts
- With dermal sinus: Dorsal dermal sinus

2) **ANTERIOR SPINAL DYSRAPHISM**

- Anterior myelomeningocele
- Anterior meningocele
- Anterior myelocele
- Anterior myelocystocele
- Anterior lipomyelocele (spinal lipoma type IC, IIC, IIIC)
- Anterior lipomyelomeningocele (spinal lipoma type VA)
- Intrasacral meningocele
- Prevertebral neurenteric cysts

3) **DYSRAPHISM CONFINED TO THE SPINAL CANAL**

- Intramedullary spinal lipoma
- Intradural lipomas
- Tight filum terminale (type IV D)
- Intrasacral meningocele
- Persistent terminal ventricle
- Retained medullary cord
- Split cord malformations
- Types 1 and 2 diastematomyelia
- Composite type diastematomyelia
- Partial type diastematomyelia
- Neuroenteric cysts
- (Epi)dermoid/dermoid
- Segmental spinal dysgenesis: type 1

4) DYSRAPHISM INVOLVING MULTIPLE ANATOMICAL COMPARTMENTS

- Dorsal enteric fistula
- Vertebral duplication
- Caudal regression syndrome
- Segmental spinal dysgenesis: type 2
- OEIS, VACTERL, Currarino's triad associations

5) NONDYSRAPHIC MASSES OF THE SPINAL REGION

- Sacrococcygeal teratoma
- Sacral chordoma

ETIOLOGY OF SPINAL DYSRAPHISM[45]

- SDs are a group of developmental disorders with multifactorial etiology, comprising genetic, environmental, and nutritional components.
- Genetic factors are probably among the most important contributing to spine developmental errors. However, despite major advances in neurogenetics over the past 10 years, little is still known of normal and abnormal spinal development. [45,46]
- Variations in some specific genes have been related to development of SD, such as MTHFR, MTHFD1, MTRR, VANGL1, VANGL2, CELSR1, and FUZ, as well as variants in the T locus on chromosome 6q. [47]
- Genes encoding proteins that participate in folate one-carbon metabolism have been broadly investigated, owing to the potential role of folic acid in preventing SD.

- The most researched gene is methylenetetrahydrofolate reductase (MTHFR), and the most widely accepted genetic component associated6 with human SD is the C677T variation of MTHFR.[48]
- Currently, there are no reliable genes or genetic counseling available for detecting human SD
- Environmental factors, such as sedentarism, poor nutrition, tobacco use, obesity in mothers, hyperhomocysteinemia, and mental stress, are key variables in this process because they are linked to excessive oxidative stress and inflammation, which shorten telomeres more quickly and accelerate maternal biologic aging.[49]

DIAGNOSTIC TESTS FOR SPINAL DYSRAPHISM

- One technique to evaluate amniotic fluid prenatally is amniocentesis. A neural-tube abnormality may be indicated by high alpha-fetoprotein (AFP) levels in the amniotic fluid. Usually, this is done between weeks 16 and 18 of pregnancy.[50]
- Another screening test is fetal ultrasonography. Fetal ultrasonography can further confirm the diagnosis in instances with high AFP. Ultrasonography can show features such as a lemon-shaped skull, a banana cerebellum, and ventriculomegaly in patients with spina bifida. By taking these assessments, caregivers can improve patient care and gain access to the right resources.[51]
- A lumbar X-ray is a useful initial screening method for observing the spine's bony architecture. It displays the degree of bone splitting and lesion.[52]
- Epidural collections, vertebral anomalies, bone septum in diastematomyelia instances, chiari malformations, related hydrocephalus, and renal anomalies that may be present in some spinal dysraphism patients are all evaluated with a computed tomography (CT) scan of the brain and spine.[53]
- The assessment of the spine, spinal cord, related abnormalities, and brain is done by magnetic resonance imaging. Owing to its excellent soft-tissue imaging, it is the most reliable technique for the diagnosis of spinal dysraphism. It is the most comprehensive research of spinal cord deformities, such as Chiari II and tethered cord.[45]

ROLE OF MRI

Yeli RK et al (2021) had included 30 patients diagnosed or provisionally diagnosed with spinal dysraphism, regardless of age or gender, based on clinical and imaging profiles. 36.67% of patients, was between one and five years old, while 33.33% were over five years old with higher (56.67%) of female population. common clinical feature was swelling in the back, observed in 14 patients, which accounted for 46.67% of the cases. Urinary incontinence and dermal sinus each affected five patients, making up 16.67%, respectively. Lower limb weakness was seen in three patients (10%), while sacral dimple was noted in two patients (6.67%). Hypertrichosis was the least common feature, affecting only one patient (3.33%). Spina bifida was the most common type, affecting 21 patients (70%). This included seven patients (33.33%) in the 0-1-year age group, eight patients (38.10%) in the 1-5-year age group, and six patients (28.57%) over five years old. ACM-II was found in eight patients (26.67%).[55]

MRI was performed on the thirty suspected cases of dysraphism by **Hosagavi RC et al**. (2019). Patients in the study ranged in age from 4 months to 11 years, with the highest number falling into the 1–5 year age group (~43.3%), predominantly with female preponderance. Lesions with subcutaneous masses (30%) were less prevalent than congenital spinal lesions without subcutaneous masses (43.3%). In patients with congenital spinal abnormalities, vertebral malformations were the most common type of spinal anomalies, followed by diastematomyelia, spina bifida, tethered cord, scoliosis, and syrinx. Spina bifida was the most prevalent spinal abnormality, accounting for 73.3% of cases. In 11 patients (36.65%), the most prevalent location of involvement was the lumbrosacral spine.[56]

Spina bifida has been identified by **Trapp B et al**. in their review as the most prevalent SD and the role of MRI being relatively reliable compared to other investigations. In their evaluation, **Santiago MR et al**. emphasized the need of MRI and CT in cases where spinal abnormalities are suspected. They studied regarding how MRI is thought to be less reliable than CT when it comes to anatomical landmarks including the height of the iliac crest, right renal artery, superior mesenteric artery, conus medullaris level, and aortic bifurcation. **Peckham ME et al**. found that it exhibited 98% accuracy in identifying the L5 vertebral body and was sufficiently accurate to identify the case of the L5 nerve root without any proximal branching.[59]

MRI can even detect transitional abnormalities at the cervicothoracic junction. The most common transitional abnormality is cervical ribs, which have been linked to lumbar sacralization. The extension of the anterior tubercle of the cervical transverse process is another lesser-known transitional anomaly that can result in the fusion of two vertebrae, usually C5 and C6.[60]

Kumar J et al in their pictorial review on MRI in SD diagnosis have explained that Myelomeningocele is the commonest common type. Also, the commonest site identified is lumbar and sacral regions accounting for 98% of all the reported cases. Eigthy percent of people with myelocele and MMC also have hydrocephalus, and all patients with Chiari II malformation—which affects the cerebellum, brain stem, base of the skull, spine, and spinal column—have the condition. Diastematomyelia is frequently associated with spinal dysraphism (SD); nevertheless, the abnormality is only called hemimyelocele when one of the hemicords has faulty neurulation. Although anterior meningocele can sometimes occur, posterior meningocele is more common in the lumbosacral area. Posterior meningocele is common in lumbosacral region but also could be seen in anterior meningocele.[61]

Another variety is terminal myelocystocele, which is characterized by the herniation of a dilated terminal central canal that forms terminal syringohydromyelia through an extended posterior vertebral defect into a dural sheath filled with CSF, which forms meningocele. It is caused by faulty secondary neurulation that alters the dynamics of CSF flow. The outer meningocele is continuous with the spinal subarachnoid space, and the inner terminal syrinx communicates with the spinal cord's central canal. Typically, the meningocele and syringocele do not communicate with one another. The few uncommon diseases identified by MRI are persistent terminal ventricle, tight filum terminale, dermal sinus, intradural lipoma, and filar lipoma. [61,62]

In 2020, **Kalabharathi H et al**. carried out a retrospective analysis using hospital databases. Sought to understand how fast MRIs relate to SD. Data from 26 newborns with myelomeningocele were included; 5 (19%) of them had spinal cord syrinxes seen on neonatal fast spine MRIs. By the time they were two years old, two patients had syrinx. Potential hazards in MRI imaging included motion artifacts and the inability to differentiate between a severe holocord syrinx and a truncated spinal cord. Thus, they came to the conclusion that rapid spine MRI obtained in the absence of sedation or anesthesia could be utilized as a screening method to identify spinal cord syrinx in newborns with SD.[63]

Tawfik NA et al (2020) had compared USG and MRI in their prospective, cross-sectional study involving 45 infants and children with suspected spinal dysraphism. They had observed an excellent agreement between MRI and ultrasound diagnosis. The accuracy of spinal USG in diagnosis of SD among ≤ 2 years old cases was comparatively inferior to MRI but not significant. The commonest anomaly was hydrocephalus in about 27% cases. Urinary incontinence among 31 (68%) of their participants. Neurological abnormalities found among 38 (84.5%) of cases. 11 (24%) had Chiari syndrome (24%).

Lumbo-sacral spine was the most common region involved in 32 patients (71.1%) followed by dorso-lumbar region.[64]

Hussein AH et al (2022) et al compared USG and MRI role in diagnosing SD. Accuracy of MRI as per their analysis was comparatively higher than USG. Rectourethral fistulas were the commonest cases they identified. Lumbosacral involvement was most frequently with incidence of 35%. Lumbar spine in 6 (30%), sacrococcygeal among 5 (25%), and two cases with dorso-lumbar. 20 (60.6%) had SD, while 13 (39.4%) with no evidence of SD.[65]

MATERIALS AND METHODOLOGY

We conducted a Prospective observational study by including all patients referred with clinical suspicion or previously diagnosed of spinal dysraphism referred for MRI SPINE to the department of Radio-diagnosis at KLE'S Prabhakar Kore Hospital and Medical Research Centre, Belagavi. The present study was conducted for the period of one year. Universal sampling method was used.

Sample size:

Formula used for sample size calculation is,

$$n = \frac{p\,(100 - p)Z^2}{E^2}$$

where n is the sample size required, p is the percentage occurrence of a state or condition (proportion or prevalence), E is the percentage maximum error required, Z is the value corresponding to level of confidence required.

Lipoma was observed in 8.57% subjects with Spinal dysraphism. Considering this at 95% confidence level and 10% maximum error, the sample size is given by,

$$n = \frac{8.57 \text{ x } (100 - 8.57) \text{ x } 1.96^2}{10^2}$$

$$n = 30.10105 \approx 30$$

Hence, minimum sample size required is 30. As sample size increases, accuracy of result also increases.

Sampling technique: Convenient sampling

Inclusion Criteria:

- All patients with clinical suspicion or previously diagnosed with spinal dysraphism irrespective of age and sex.

Exclusion Criteria:

1. Patients with history of claustrophobia
2. Contraindications to MRI
3. Patients with history of metallic implant insertion, cardiac pacemakers and metallic foreign body insitu.
4. Patients not willing to take part in the study.

Study protocol:

Study was taken only after obtaining the ethical clearance from the institutional ethics committee. Patients were included based on the above-mentioned inclusion and exclusion criteria. Written informed consent was obtained from those patients aged more than 18 years and the consent from parents of children after explaining the procedure in their understandable language.

Study was done using a 3.0 Tesla Siemens MRI machine (Mangnetom Spectra). Standard scan protocol was done for all the patients with suspected or previously diagnosed case of spinal dysraphism. MR myelography protocol was used in all patients. A pre-structured proforma was used for collection of relevant demographic data, family and gestational history & clinical data. The above-mentioned study population were subjected to a study of MRI spine on 3.0 Tesla Siemens MRI machine (Mangnetom Spectra). Routine MRI spine protocol and MR myelography protocol is used.

STATISTICAL METHOD

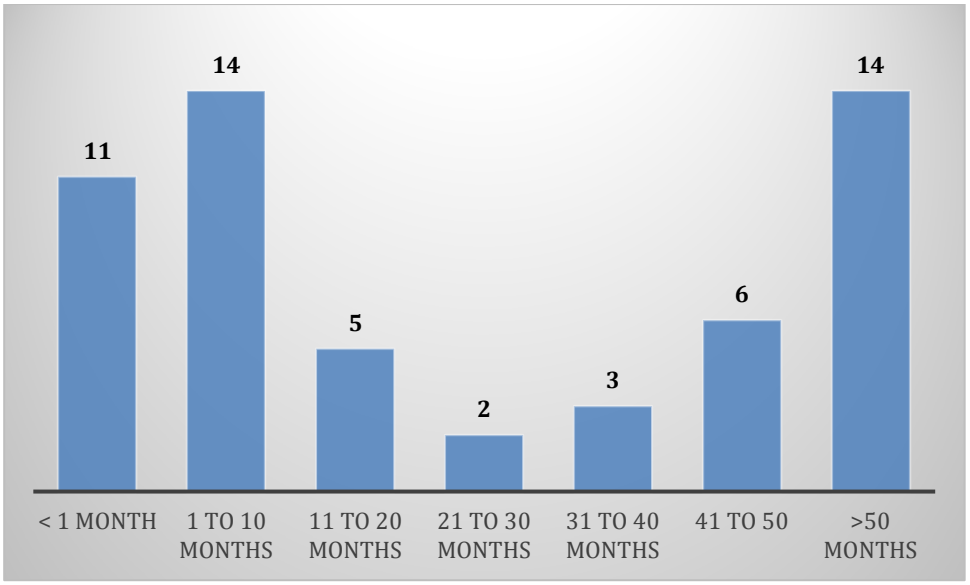
Data was analyzed using statistical software R version 4.2.0 and Microsoft Excel. Categorical variables were represented by frequencies and percentages. Continuous variables were represented by Mean ± SD / Median (Min, Max) form. Only descriptive analysis was applicable for the parameters we had taken as there was neither the comparison group nor the controls.

RESULTS

Table 1: Distribution of age

Age in month	**N**	**%**
</=1 month	11	20.0%
1.1 to 12 months	17	30.9%
12.1 to 24 months	4	7.3%
24.1 to 36 months	2	3.6%
36.1 to 48 months	6	10.9%
48.1 to 60 months	3	5.5%
>60.1 months/ 5 years	11	20.0%
Average age	29.91±8.6 months	
Minimum	0.5 month	
Maximum	96 months	

Out of 55 patients, those aged between 1.1 to 12 months were more with incidence of 30.9% (17/55). Whereas the average age was 29.91±8.6 months.

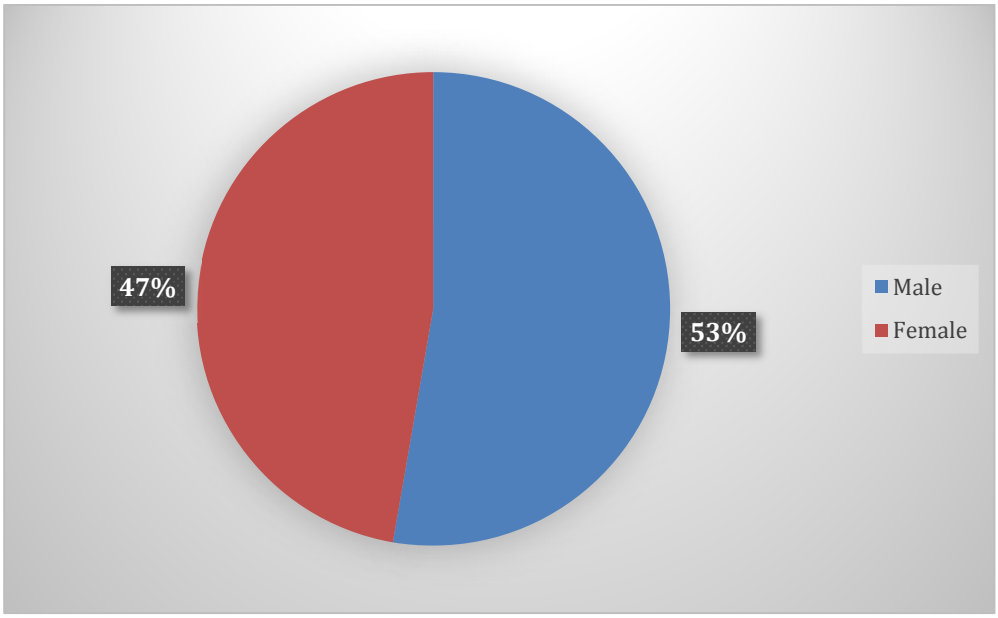


Graph 1: Distribution of age

Table 2: Distribution of gender and presence of SD

Gender	**N**	**%**
Male	29	52.7%
Female	26	47.3%

29 of 55 (52.7%) were males and 26 (47.3%) were females.

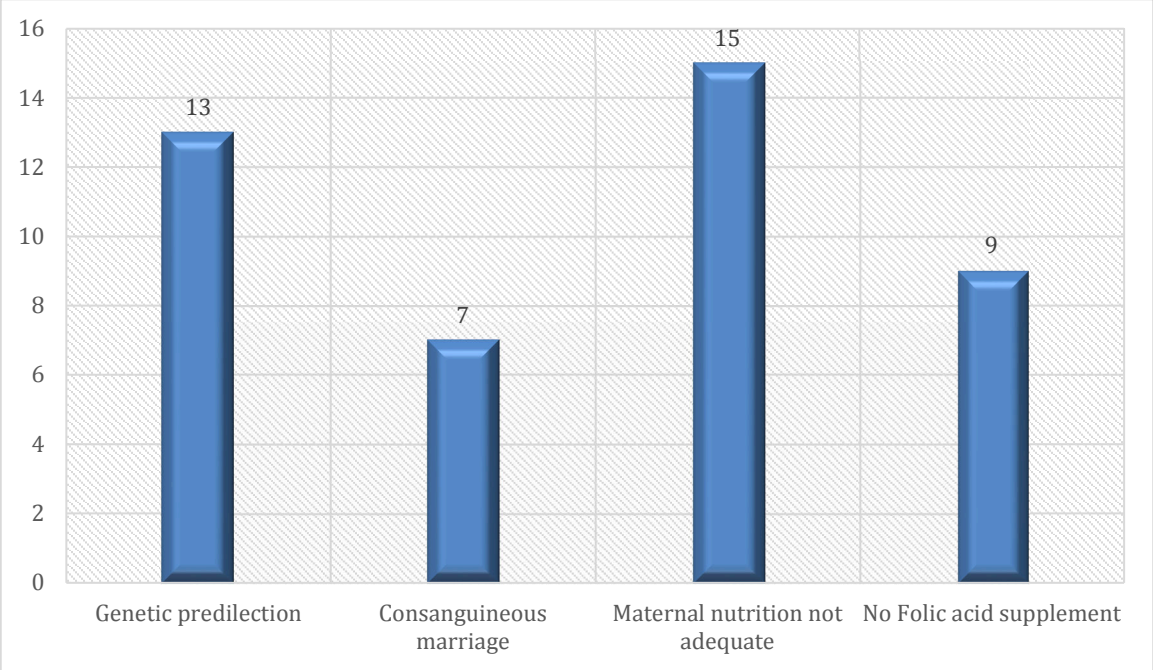


Graph 2: Distribution of gender

Table 3: Associated history

Parameter	N	%
Maternal nutrition not adequate	15	27.3%
Genetic predilection	13	23.6%
No Folic acid supplement	9	16.4%
Consanguineous marriage	7	12.7%

We can observe that the maternal inadequacy of the nutrition was being the commonest predisposing factor, accounting for about 27.3% (15) followed by 13 (23.6%) with genetic predilection.

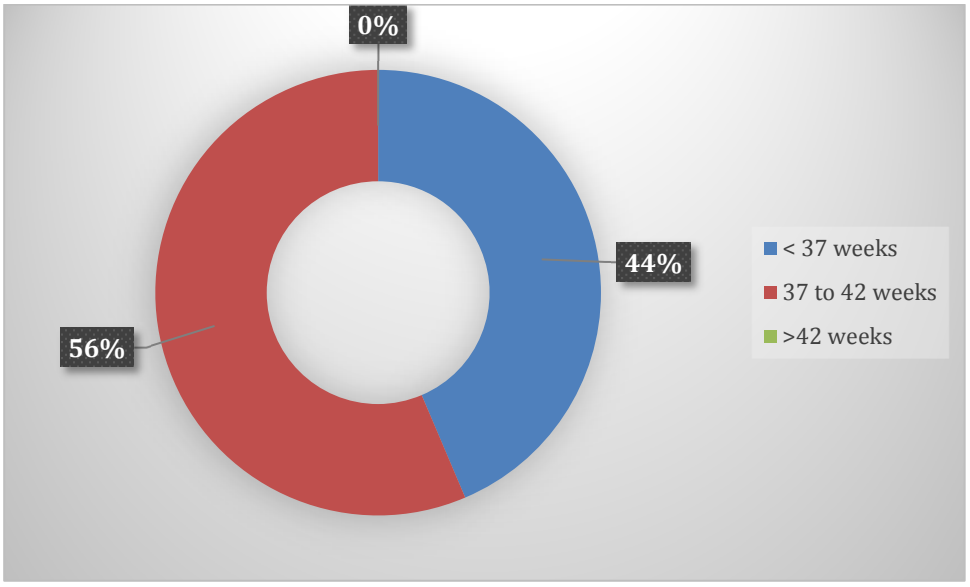


Graph 3: Distribution of the patients based on associated factors for SD

Table 4: Gestational age at birth

Gestational age in weeks	N	%
<37 weeks	24	43.6%
37 to 42	31	56.4%
>42 weeks	0	0

Majority of them had delivered at normal gestational age at birth whereas 24 (43.6%) had pre term labour.

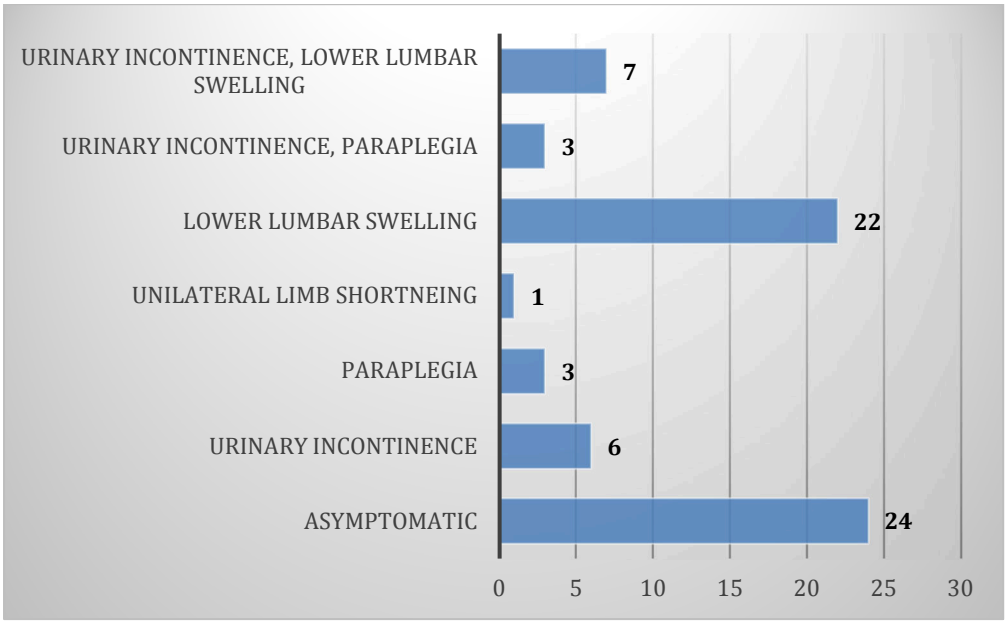


Graph 4: Distribution of study population based on the gestational age at the time of birth

Table 5: Distribution of symptoms for SD

Symptoms	N	%
Asymptomatic	24	43.6%
Lower lumbar swelling	22	40.0%
Urinary incontinence, Lower lumbar swelling	7	12.7%
Urinary incontinence	6	10.9%
Paraplegia	3	5.5%
Urinary incontinence, Paraparesis	3	5.5%
Unilateral limb shortening	1	1.8%

Lower lumbar swelling was the commonest symptoms accounting for about 22 (40%) of the study population.

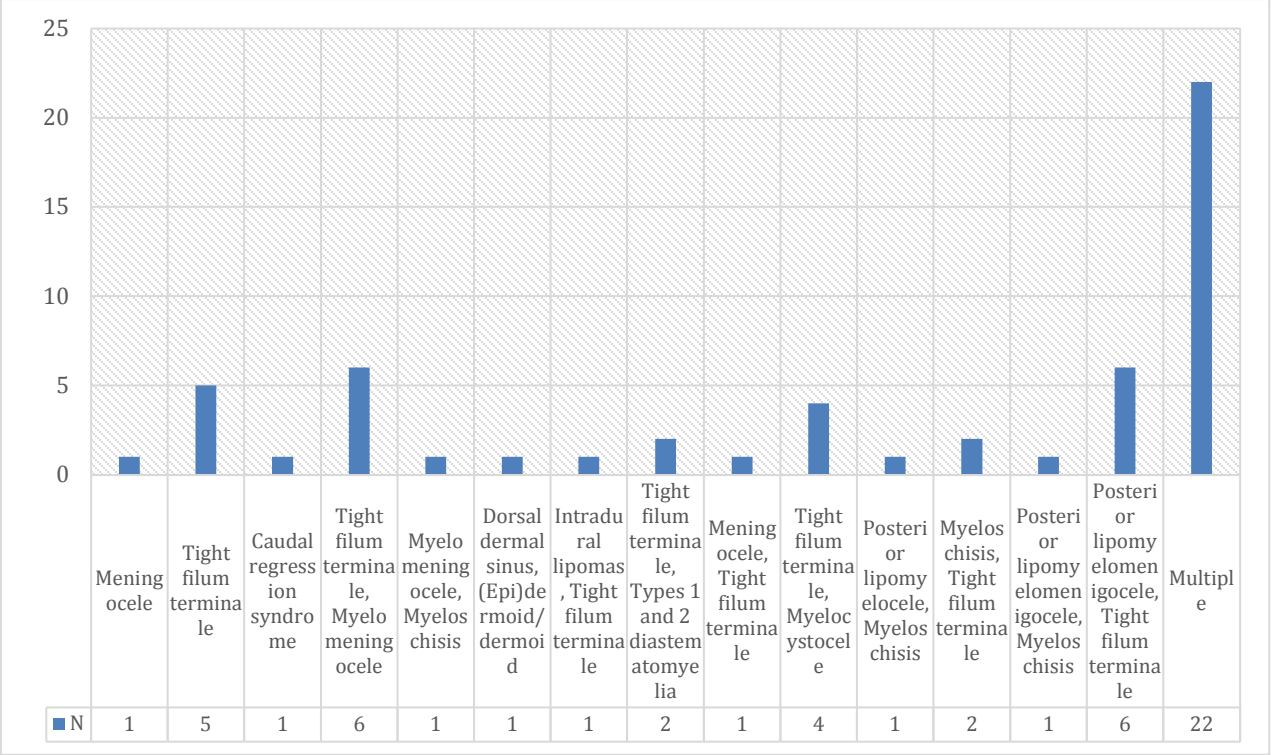


Graph 5: Distribution of symptoms

Table 6: Type of SD

Parameter	N	%
Multiple	22	40.0%
Tight filum terminale, Myelomeningocele	6	10.9%
Posterior lipomyelomenigocele, Tight filum terminale	6	10.9%
Tight filum terminale	5	9.1%
Tight filum terminale, Myelocystocele	4	7.3%
Tight filum terminale, Types 1 and 2 diastematomyelia	2	3.6%
Myeloschisis, Tight filum terminale	2	3.6%
Meningocele	1	1.8%
Caudal regression syndrome	1	1.8%
Myelomeningocele, Myeloschisis	1	1.8%
Dorsal dermal sinus, (Epi)dermoid/dermoid	1	1.8%
Intradural lipomas, Tight filum terminale	1	1.8%
Meningocele, Tight filum terminale	1	1.8%
Posterior lipomyelocele, Myeloschisis	1	1.8%
Posterior lipomyelomenigocele, Myeloschisis	1	1.8%

Majority of the patients had multiple defects observed on MRI and few were with single and two different anomalies which are represented in the above table and same is depicted below.

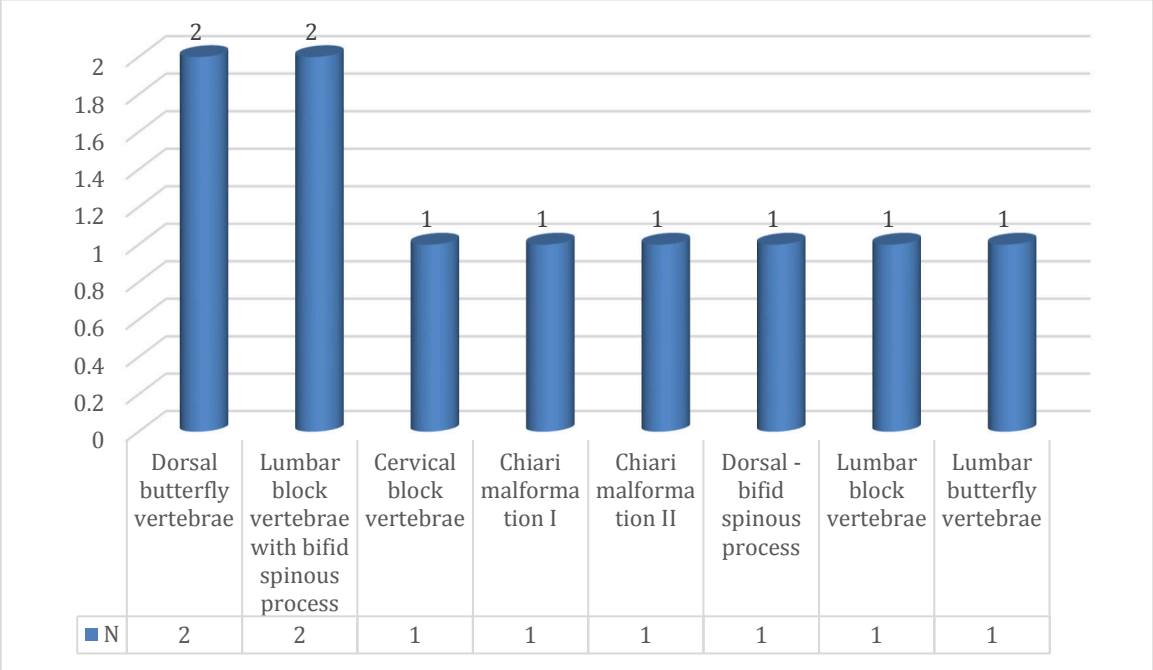


Graph 6: Type of SD observed on MRI

Table 7: Other associated vertebral anomalies

Associated vertebral anomaly	**N**	**%**
Dorsal butterfly vertebrae	2	3.6%
Lumbar block vertebrae with bifid spinous process	2	3.6%
Cervical block vertebrae	1	1.8%
Chiari malformation I	1	1.8%
Chiari malformation II	1	1.8%
Dorsal -bifid spinous process	1	1.8%
Lumbar block vertebrae	1	1.8%
Lumbar butterfly vertebrae	1	1.8%
Total	**10**	**18.88%**

10/55 were presented with other associated changes which are described in the above table and below graph.



Graph 7: Associated vertebral anomalies

Table 8: Distribution of study population based on associated renal anomaly

Renal anomaly type	**N**	**%**
Bilateral moderate hydronephrosis	2	3.6%
Ectopic left kidney	3	5.5%
Total	5	9.1%

DISCUSSION

We had aimed at analysing the distribution pattern of SD changes on MRI among the suspected cases of SD. We had included 55 patients irrespective of the age and gender. Those aged between 1.1 to 12 months were more with incidence of 30.9% (17/55) followed by 20% (11/55) each aged </=1month and > 5 years. 6 (10.9%), 4 (7.3%), 3 (5.5%) and 2 (3.6%) were aged between 36.1 to 48 months, 12.1 to 24 months, 48.1 to 60 months and 24.1 to 36 months respectively. There was statistically significant association with age or gender distribution observed. Similarly, in **Trapp et al** the average age of their patients with SD was 15.6 ± 13 months. Unlike our study, patients with age group 2 years were more whereas our study had equal distribution of those aged more and less than 5 years.[57] This was even same outcome as observed by **Kumari et al.**[66] Unlike our findings, **Dhingani D et al** had broadly mentioned that the patients aged <10 years being higher. This might be due to the concern of symptoms among the children noticed by parents which would led them for clinical evaluation.[67]

Maternal inadequacy of the nutrition was being the commonest predisposing factor, accounting for about 15 (27.3%) followed by 13 (23.6%) with genetic predilection. 9 (16.4%) had not taken folic acid supplements and 7 (12.7%) had history of Consanguineous marriage of their parents. 24 (43.6%) were pre term labour at birth and 31 (56.4%) were born at normal gestational age. No history of post-dated delivery was observed.

We found that 43.6% were asymptomatic. 22 (40%) had Lower lumbar swelling as their symptom followed by 7 (12.7%) with urinary incontinence with lower lumbar swelling. 6 (10.9%) had only urinary incontinence. Three each were presented with Paraplegia and Urinary incontinence with paraparesis. Unilateral limb shortening was observed among only one patient. Even among **Kumari MV et al,** [66] **Dhingani D et al**[67] and **Mehta DV et al,** [68] the commonest symptom was swelling over lower back, respectively. Unlike the present assessment, **Dhingani D et al** had even found hair tuft, dermal sinus, incontinence of feces as the major worrying symptoms in their study population but they also did not find any significant association of these with demography or the clinical history.[67] **Kumar J et al** also depict the same in their interpretation, mentioning as lumbar region involvement being the commonest.[67]

So, we could interpret that as the embryological reviews, the spinal deformity either in open or in closed type, the bulge in lower lumbar has been the commonest persistent finding irrespective of the age.

We found that there rarely the similar type of SD changes observed. Majority of them were with multiple changes/defects in the vertebra and spine accounting for about 22 (40%). Of all, tight filum terminale with myelomeningocele and tight filum terminale with posterior lipomyelomenigocele were the commonest co-existing changes with the incidence of 10.9% (6/55). Of all the existing changes, tight filum terminale and myelomeningocele were the commonest. Also, there was no specific anterior or posterior SD changes observed. It was mainly the association of both together.

On further analysis, we could observe that dorsal butterfly vertebrae and Lumbar block vertebrae with bifid spinous process were observed among two each of our patients as another associated anomalies. One each were observed with Cervical block vertebrae,

Chiari malformation I, Chiari malformation II, Dorsal -bifid spinous process, Lumbar block vertebrae and Lumbar butterfly vertebrae respectively. In Mehta DV et al, type II Arnold-Chiari malformation with lumbar meningomyelocele, spina bifida occulta and diastmatomyelia were the commonest with the incidence of 34%, 22% and 18% respectively. Below are the MRI from few of our study population. Thos was same as **Tawfik NA et al** findings.[64]

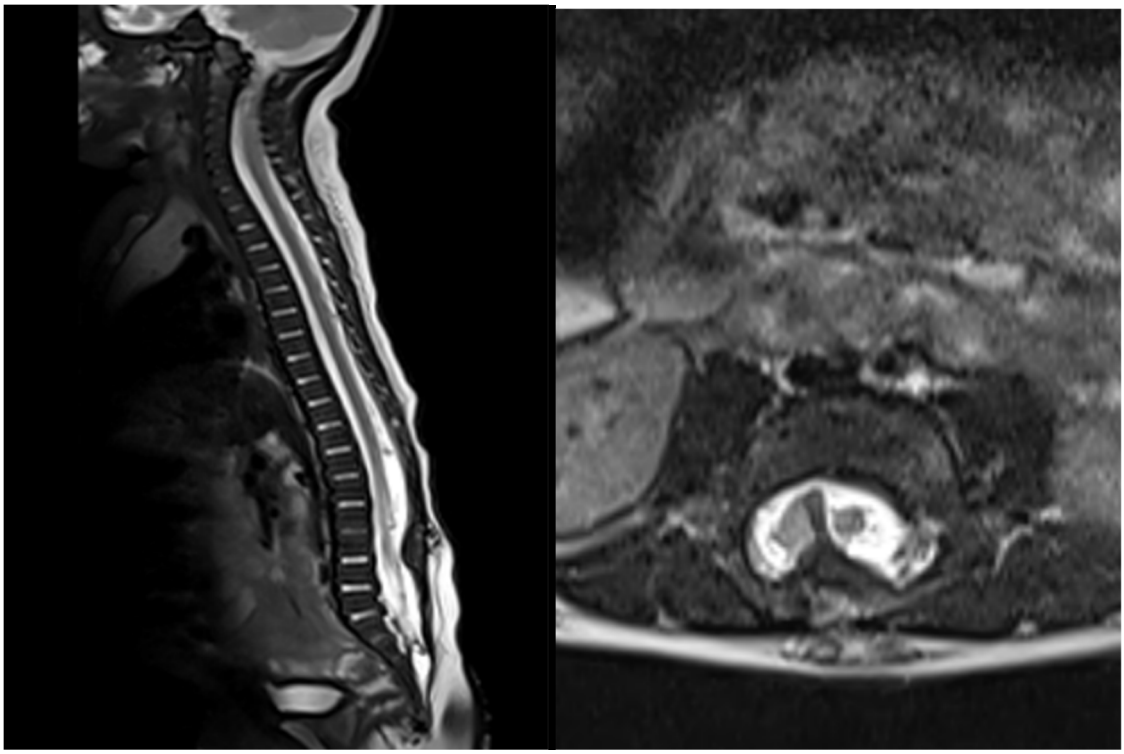


FIGURE 6 : **Case 1 -** These are the MRI findings from a 12-month-old male patient came with complaints of lower lumbar swelling since birth. MRI sagittal and axial views of the spine shows a diastometamyelia in the lumbar region at the level of L4 vertebra and extending below at the level of L5 and fusing at level of superior end plate of S1 vertebra with separation of the spinal cord by thin septa. Low lying spinal cord with the conus medullaris ending at the level of S3 vertebral body. Hydrosyringomyelia below L1-L3 level. Dilatation of the terminal central canal suggestive of ventricularis terminalis. Closed spinal dysraphism with neural placode and meninges seen posteriorly outside the spinal canal in the subcutaneous plane of lumbar region at the level of L3-L4 vertebrae. **Features are suggestive of closed posterior spinal dysraphism with lipomeningocele and complete diastometamyelia.**

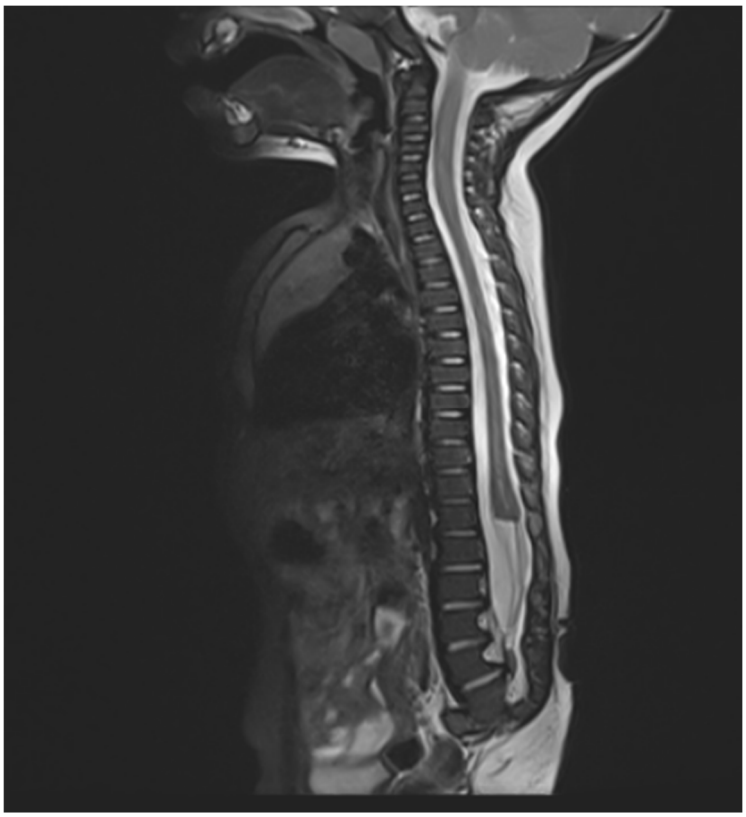


FIGURE 7: Case 2 - 7-month-old male patient was referred from MRI spine. T2w sagittal image of spine - Non visualization of all the sacral vertebrae & coccy agenesis. The spinal cord is seen to terminate at the level of midportion of D12 vertebral body. The conus medullaris is blunt and terminates at the lower border of D12 vertebral body with truncated appearance of conus medullaris. Dilated central canal from D11 to D12 vertebral bodies. Tethering of the spinal cord noted at the level of L4 vertebra. **Features are suggestive of caudal regression syndrome (Group I).**

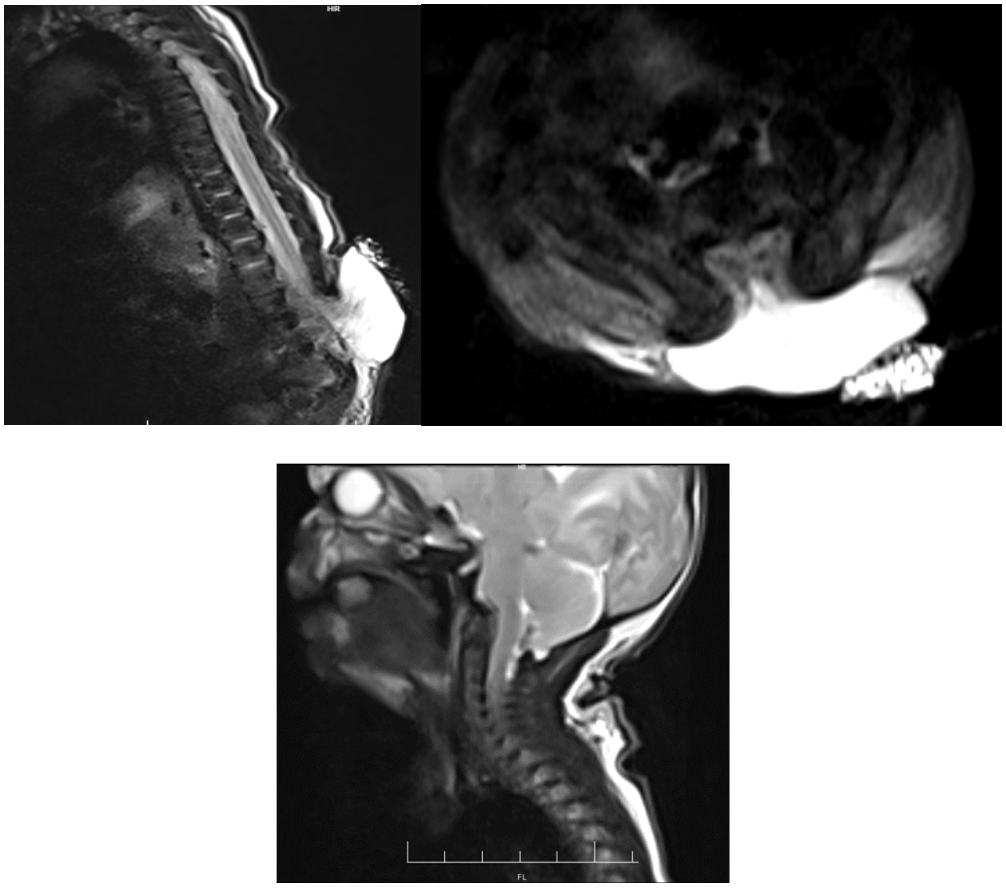


FIGURE 8 : Case 3- These are the MRI from a 2 day old male patient came with complaints of lower lumbar swelling, on MRI spine, T2w sagittal and axial images there is seen a defect along the posterior vertebral element at L5, S1 and S2 vertebral levels with herniation of thecal sac, CSF and neural elements posteriorly into the extra-vertebral space and reaching upto the subcutaneous plane suggestive of meningomylocele. There is a defect along the superior aspect of meningomylocele sac with herniation of CSF and few of the neural elements likely suggestive of rupture of meningomylocele. Peg like herniation of cerebellar tonsils below the foramen magnum into the upper cervical canal suggestive of Chiari II malformation. **Features suggestive of lumbosacral meningomyelocele with a defect in the myelomeningocele sac with rupture.**

Irrespective of any given clinical study, either the retrospective or the prospective we did not find much association between the distribution of specific type of SD, almost all cases were presented with multiple anomalies than single specific. **Barnes KS et al** who had assessed for SD changed by using various imaging modalities as well observed the similar outcome.[69]

Bilateral moderate hydronephrosis among two cases and left ectopic kidney in one patient were the associated renal anomalies. Ruptured extra spinal collection was found among one 15 days old baby. As per the available evidences, caudal regression would be associated with renal anomalies. Also, the various other SD changes have been found to be associated with other renal changes. Either the symptoms or the type of SD and even vertebral changes, associated renal anomalies did have any specific pattern of distribution. To substantiate this, we can quote **Hussein AH et al** who had observed rectourethral fistulas among their study population.[65]

LIMITATIONS:

With the above discussion, we could assess that there is no correlation between the demographic data, clinical history, and presentation of SD. This could be due to almost all of patients being presented with closed type of SD among whom the symptoms were no major and also the complications were not persistent. As we did not include any control group or the operative findings, we could not assess for the diagnostic accuracy of MRI. Which is the major limiting factor.

CONCLUSION

In a study population of 55, the average age of our patients who presented with spinal dysraphism was 29.91±8.6 months. Minimum age being 0.5 month (15 days) and maximum was 96 months. 29 (52.7%) males and 26 (47.3%) females were observed. Maternal malnutrition was the commonest predisposing factor (27.3%) followed by Genetic predilection among 23.6%. Lower lumbar swelling among 40% (22 cases) was the most common symptom. Patients with multiple spinal and vertebral changes were more accounting for 40% of the cases. 18.88% had associated vertebral deformities. 5 (9.1%) were found with renal anomaly.

SUMMARY

Congenital conditions that cause aberrant growth in the spine and/or spinal cord are referred to as spinal dysraphism. The commonest forms are myelomeningocele, meningocele, lipomeningocele, lipomyelomenigocele, rachischisis and myeloschisis.

Adequate folic acid consumption throughout pregnancy has been demonstrated to dramatically lower the incidence of myelomeningocele patients. Pregestational diabetes may raise the probability of spinal dysraphism and other abnormalities of the central nervous system. Nutritional deficiency accounts for up to 50% of all cases of spinal dysraphism, making it the most often reported risk factor.

There are 0.5 to 8 incidences of both open and closed spinal dysraphism for every 1,000 live births.

We conducted a Prospective observational study by including all patients referred with clinical suspicion or previously diagnosed of spinal dysraphism referred for MRI SPINE to the department of Radio-diagnosis at KLE'S Prabhakar Kore Hospital and Medical Research Centre, Belagavi.

The objective of the study was to characterize and categorize the site and type of spinal dysraphism as seen on magnetic resonance imaging (MRI).

The sample consisted of 55 patients with spinal dysraphism. Exclusion criteria were patients with claustrophobia, any patient with a contraindication for MRI.

After obtaining informed consent, baseline data was recorded on a self-designed proforma. MRI spine was performed on 3.0 Tesla Siemens MRI machine (Mangnetom Spectra). We characterized and classified the spinal dysraphisms according to the new anatomical-clinicoradiological classification.

We observed that the maternal inadequacy of the nutrition was the commonest predisposing factor, accounting for about 27.3% followed by 23.6% with genetic predilection. In our study mean age was 29.91±8.6 months, maternal inadequacy of the nutrition was being the commonest predisposing factor, lower lumbar swelling was the commonest symptoms.

Tight filum terminale was the most common of the spinal dysraphisms seen. SD was associated with renal and vertebral anomalies. Complications of SD as observed as extraspinal collection, rupture of the myelomeningocoele was observed in a few cases.

Most of the closed spinal dysraphisms remain asymptomatic during birth and can be suspected only in the presence of cutaneous markers or clinical manifestation of neurological deficits in later stages of life. MRI has facilitated in both diagnosis and classifying of these disorders and decision making of earlier &individually tailored treatment.

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ANNEXURE I –

INFORMED CONSENT

"ROLE OF MRI IN CHARACTERIZATION AND CATEGORIZATION OF THE SITE AND TYPE OF SPINAL DYSRAPHISM – A ONE YEAR HOSPITAL BASED OBSERVATIONAL STUDY"

Name of Student/Principal Investigator: REG NO.- BS0121013

Objective: The objective of this study is to characterize and categorise the site and type of spinal dysraphism as seen on magnetic resonance imaging (MRI).To study the additional/ associated findings in cases of spinal dysraphism.

Introduction: Your child/ward is being invited to participate in this study to categorise and characterize the spinal dysraphism using MRI spine.

Explanation of procedure: If, you agree to be part of the research study, you will be asked the relevant history and your child/ward will be subjected to relevant clinical examination and investigations.

Withdrawal from participation in the study: Participation in this study is voluntary. You will be free to decide whether to participate in this study or continue participation once enrolled. In case you decide to withdraw your participation, you are free to do so. However, please convey the decision to the principal investigator.

Possible benefits from participating in the study: You will not have nor get any benefits by participating in this study. The data gathered will help the population at large.

Possible risks from participating in the study: There are no risks involved in participating in this study.

Privacy and confidentiality: The information collected from you will be coded, to prevent any person from identifying you. Your identity will never be revealed. The data collected from you will be kept confidential and only processed or aggregated data will be used for publication.

Financial incentives: You will not receive any payment for participating in this study.

Authorization for publication of aggregated data: Results obtained after processing of the aggregated data will be published for scientific purposes and or presented to scientific groups. However, your identity will never be revealed.

Questions: In case of any questions with regard to this study, you are free to contact:

Registration number- BS0121013

Postgraduate

Department of Radiodiagnosis

Jawaharlal Nehru Medical College KAHER,

Belagavi – 590010 Karnataka

Dr.____

Professor,

Department of Radiodiagnosis,

Jawaharlal Nehru Medical College,

KAHER, Belagavi - 590010 Karnataka

Legal rights: By signing this consent form, we are not waving any of your legal rights.

CONSENT STATEMENT

I am making a voluntary decision to participate in the study "**ROLE OF MRI IN CHARACTERIZATION AND CATEGORIZATION OF THE SITE AND TYPE OF SPINAL DYSRAPHISM – A ONE YEAR HOSPITAL BASED OBSERVATIONAL STUDY**". My signature below indicates that I have decided to participate and I have read the information provided above or the information provided above has been read to me in the language that I understand best. I was given the opportunity to ask questions and that they have been answered to my satisfaction.

Name of the participant:

Signature or left thumb impression of the parent/participant:

Name of the witness:

Signature or left thumb impression of the witness:

Name of the investigator:

Signature of the investigator:

ANNEXURE II–

PROFORMA FOR DATA COLLECTION

NAME_____

AGE_____ **OP/IP NO** _____

MOBILE_____

ADDRESS _____

CHIEF COMPLAINTS:

HISTORY OF PRESENTING ILLNESS

ANTENATAL HISTORY:

BIRTH HISTORY:

GESTATIONAL AGE AT BIRTH:

PAST HISTORY

FAMILY HISTORY

INVESTIGATION:

INTERVENTION- NIL

CATEGORISATION OF SPINAL DYSRAPHISM:

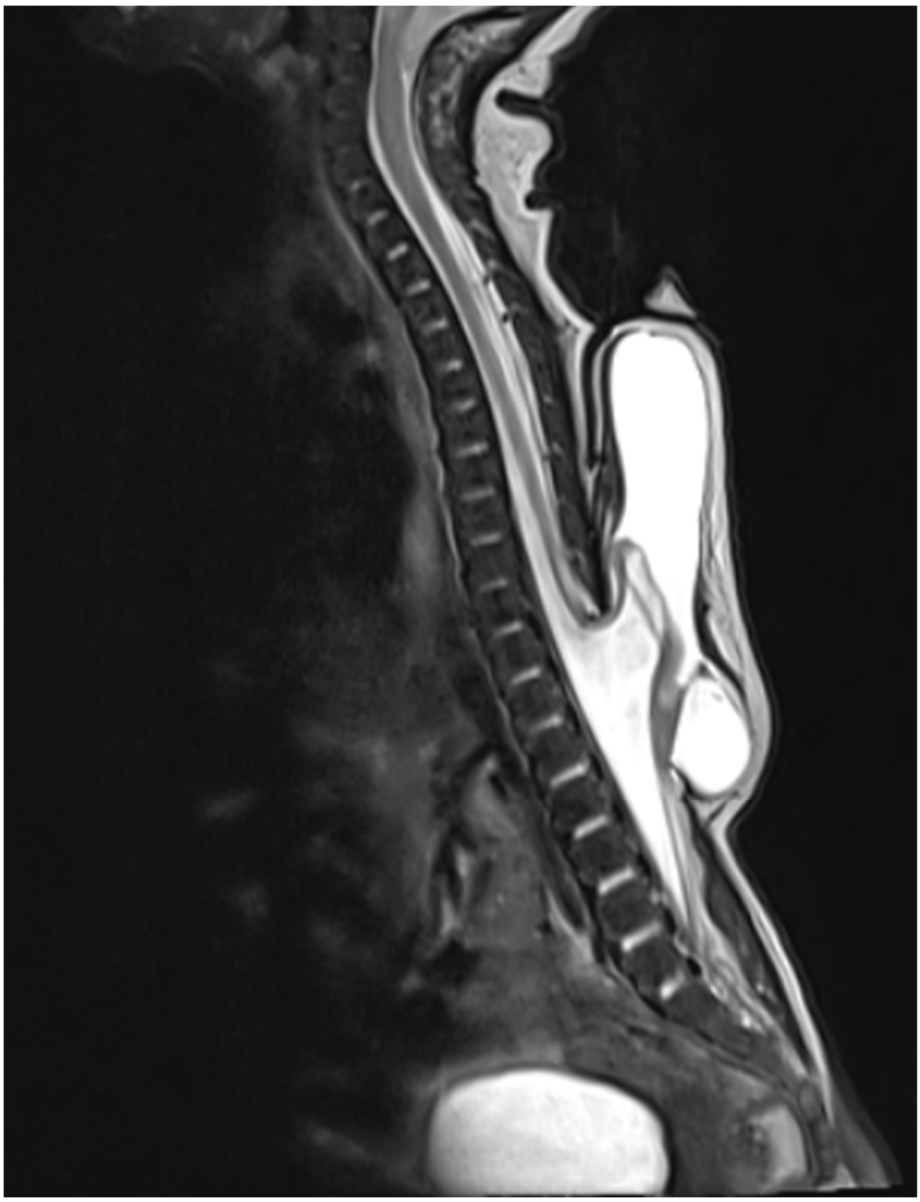
1) **Posterior spinal dysraphism** **Open spinal dysraphism**: **Closed spinal dysraphism:** ➢ **With subcutaneous mass:** ➢ **Without subcutaneous mass (cutaneous stigmata if any)**	
2) **Anterior spinal dysraphism**:	
3) **Dysraphism confined to the spinal canal:**	
4) **Dysraphism involving multiple anatomical compartments:**	
5) **Nondysraphic masses of the spinal region**	

CHARACTERISATION OF SPINAL DYSRAPHISM:

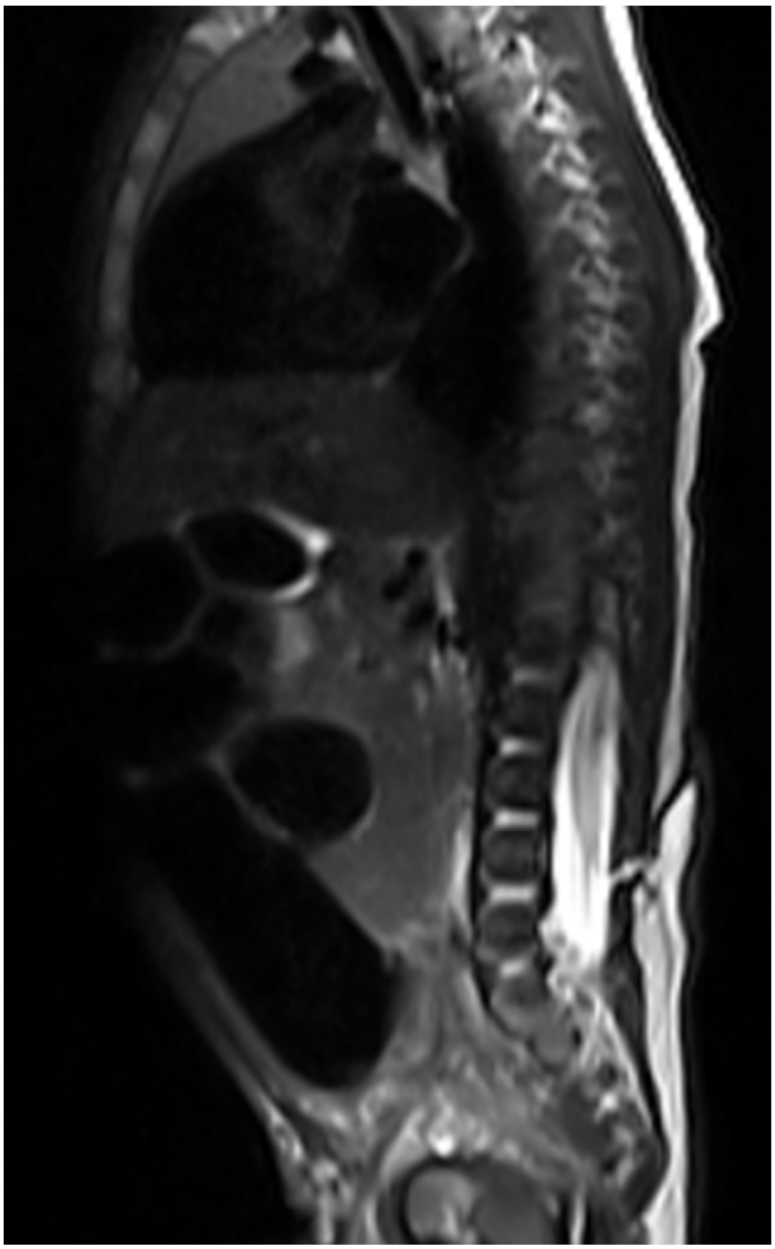
ADDITIONAL/ ASSOCIATED FINDINGS ASSOCIATED WITH SPINAL DYSRAPHISM (IF ANY):

ANNEXURE III: FIGURES

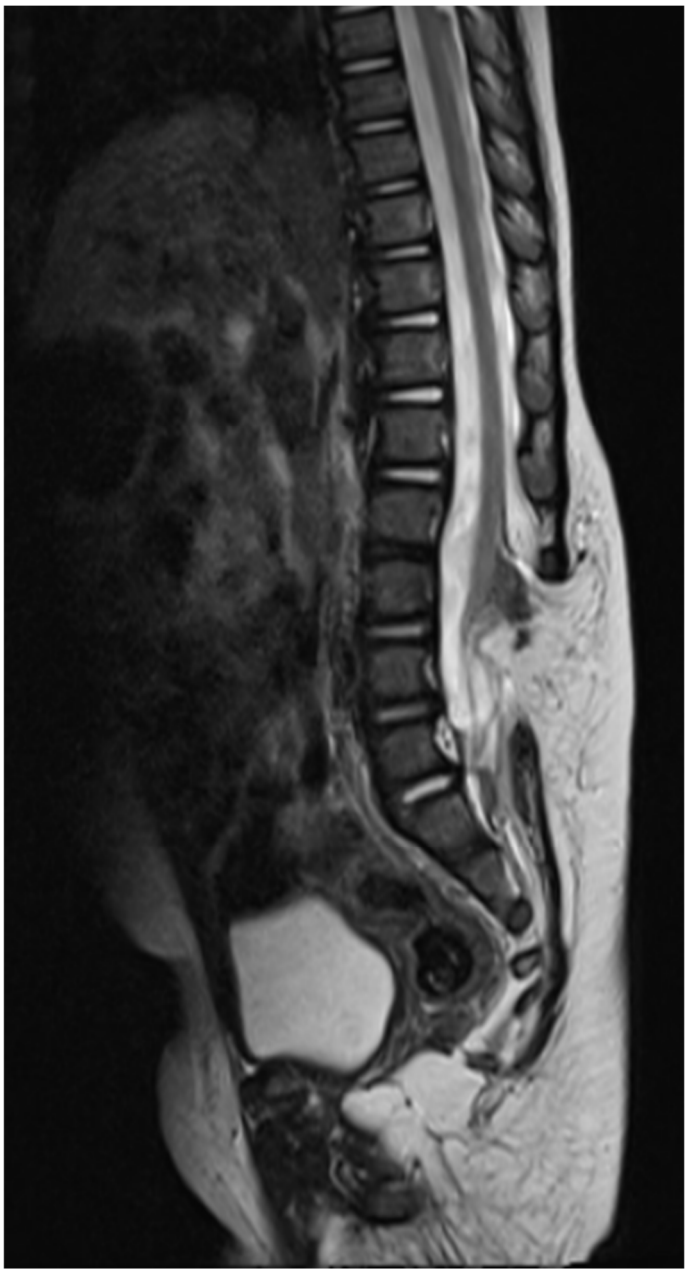
3.0 Tesla Siemens MRI machine (Mangnetom Spectra) used for the study



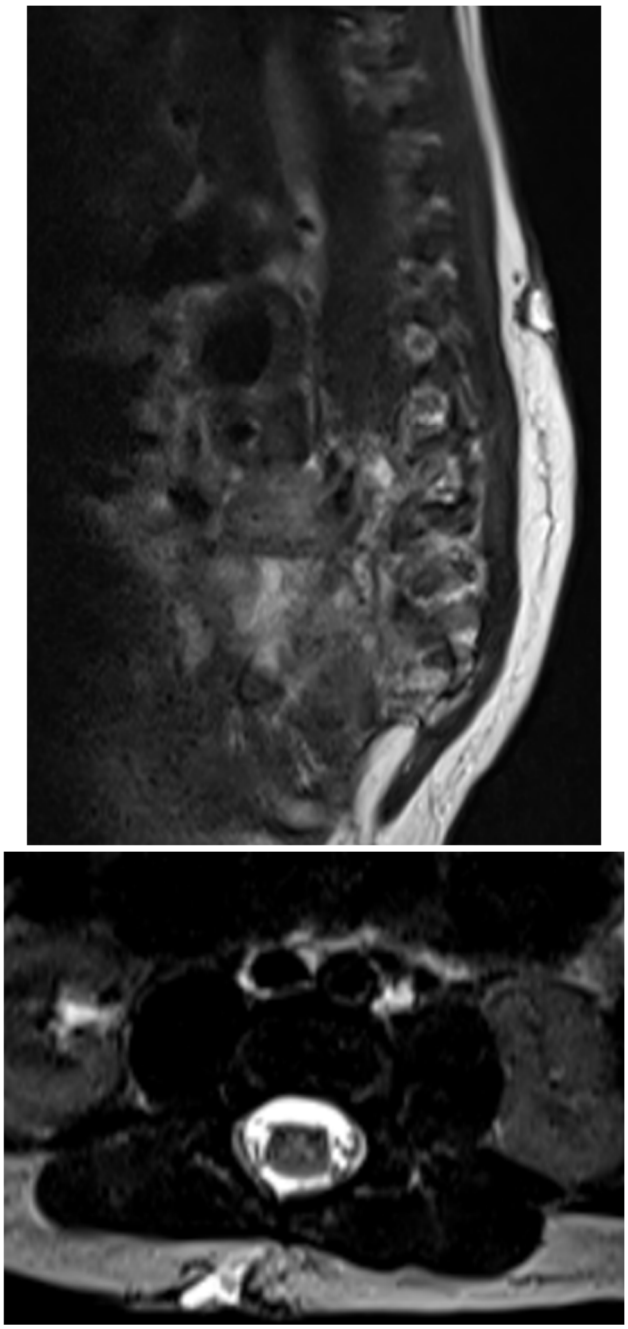
1. 14 day old female patient came with complaints of lower lumbar swelling since birth, T2w MRI sagittal image shows outpouching of thecal sac through a midline defect at the level of D11 to L2 vertebral bodies with herniation of CSF, meninges and neural placode along the dorsal aspect of thoracolumbar spine. Spinal cord is tethered to the myelomeningocele. Peg like herniation of cerebellar tonsils below the foramen magnum suggestive of cerebellar tonsillar herniation. **Features suggestive of dorsal meningomyelocele at thoraco lumbar region.**



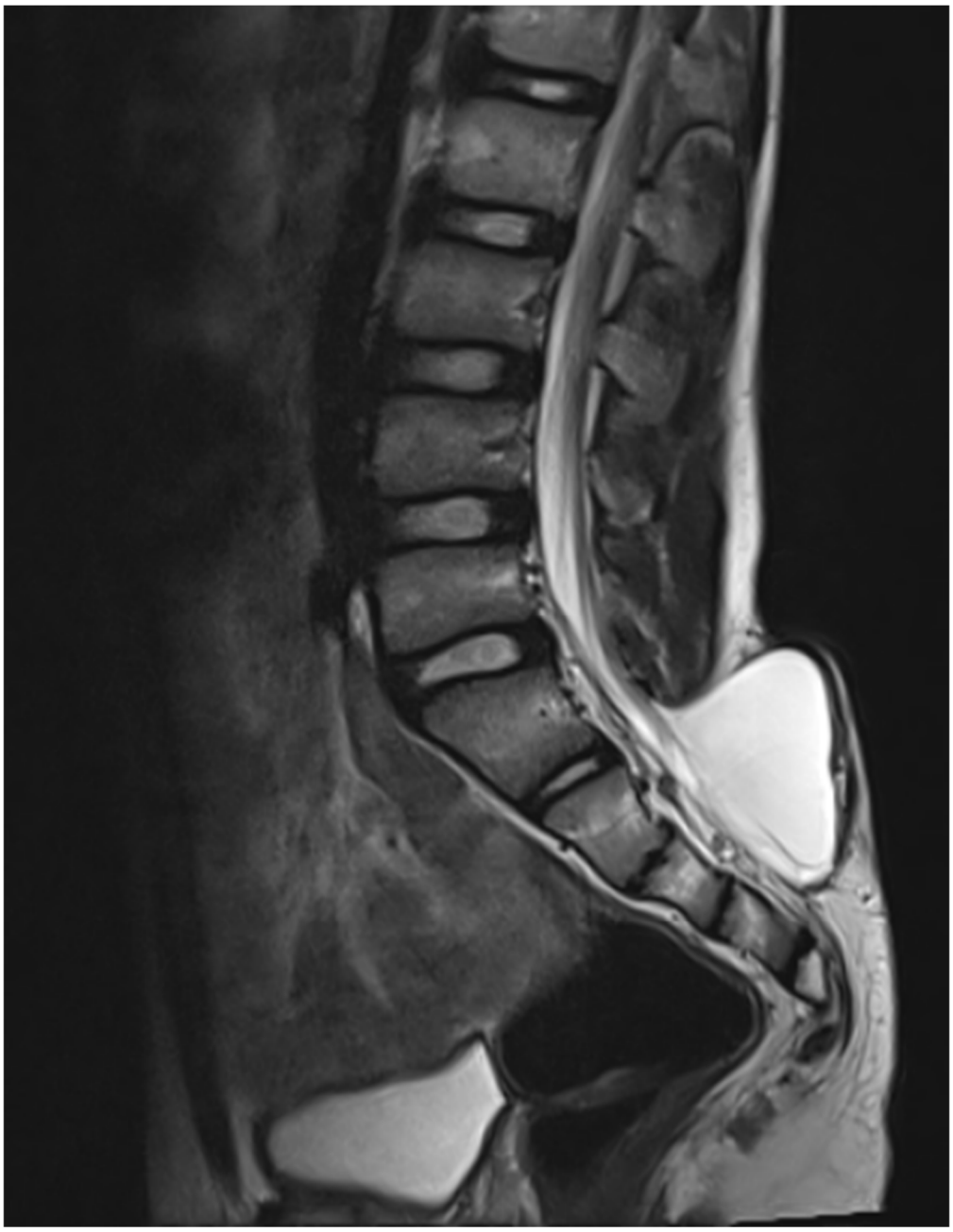
2. 3 day old male patient came with complaints of lower lumbar swelling since birth, on MRI spine T2w sagittal image, widening of distal cord in the conus from the level of lower border of L2 to upper border of L4 vertebra with herniation of CSF filled thecal sac posteriorly into the subcutaneous plane suggestive of meningocele. Suspicious tract arising from the thecal sac which is seen communicating with an extra-spinal collection. **Features suggestive of Meningocele with a tract communicating with an extra-spinal collection.**



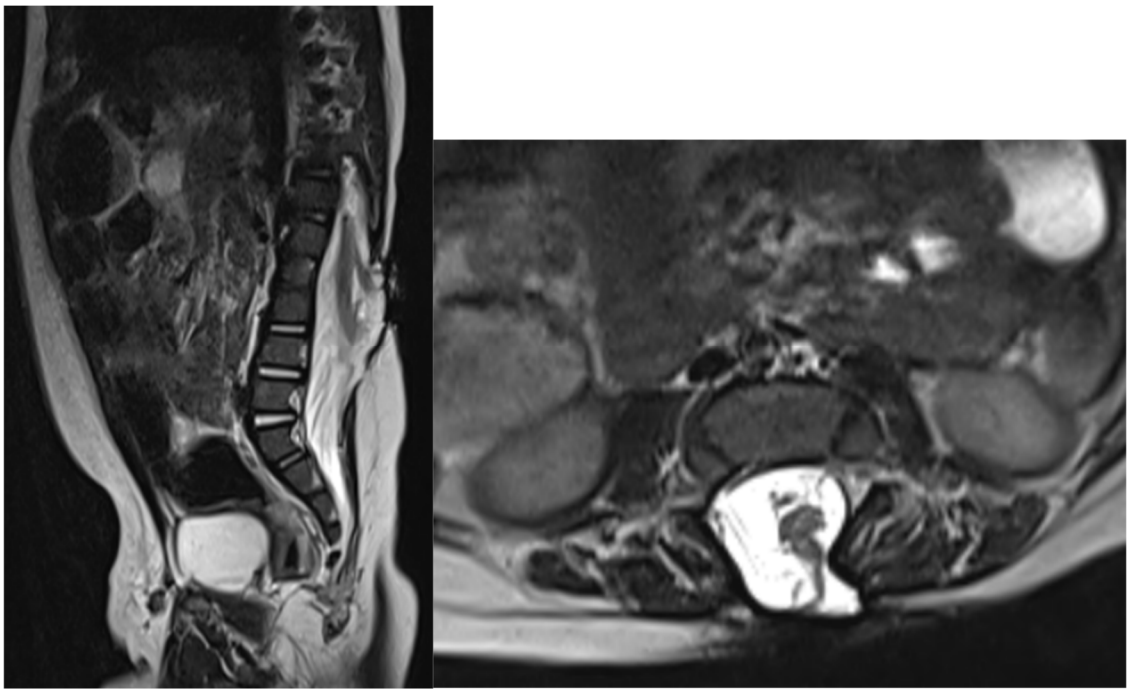
3. 12 month old female patient came with complaints of lower lumbar swelling since birth, MRI spine sagittal image shows widening of distal cord in the conus from the level of upper border of L3 to L4 vertebra with a fatty mass dorsal to the cord at L3-L4 level and is seen to be in contiguous with subcutaneous fat through a bony defect posteriorly with herniation of a few of the neural elements through it. Low lying spinal cord with the conus medullaris ending at L4 vertebral body. Tethering of the neural elements to the thecal sac at L4 level. **Features suggestive of Lipomyelocele with low lying conus and tethered cord.**



4. 9 month old female patient was referred for MRI spine. T2w MRI images show partial splitting of the spinal cord (diastematomyelia) in the lumbar region, extending from L1 vertebra up to superior end plate of L3 vertebra. Low lying cord with conus medullaris lying at L4 vertebra with tethering of nerve roots to the dural sac. Linear T2 hyperintense tract extending from the surface of dermal lesion into the thecal sac at the level of L3 vertebra. **Features are suggestive of Diastematomyelia , Low lying cord with conus medullaris at L4 vertebra, and dorsal dermal sinus.**



5. 10 year old male patient was referred for MRI spine. T2w sagittal image shows outpouching of thecal sac through the unfused posterior elements from the level of S1 to S4 vertebral bodies, with herniation of CSF, meninges and cauda equina suggestive of myelomeningocele **Features suggestive of Sacral myelomeningocele.**



6. Conus is ending at L5 vertebral body suggestive of low lying cord. Widening of the spinal canal in the lumbosacral region with nerve roots adherent to the thecal sac suggestive of tethered cord. CSF filled outpouching of the thecal sac through the defect in the posterior arch of the vertebral canal from upper border of L2 to lower border of L4 vertebrae with herniation of the neural elements into it suggestive of meningomylocele. Splitting of the spinal cord from lower border of L1 vertebral body with one the hemi- cord and its nerve roots entering into the CSF filled outpouching suggestive of diastometamyelia [Hemimyelomeningocele]. Thinning of the spinal cord with central T2 cord hyperintensity from C6 to D12 vertebral body suggestive of syrinx**. Features suggestive of Lumbar hemimyelomeningocele with low lying conus, tethered cord and diastometamyelia.**

ANNEXURE IV: KEY TO MASTERCHART

I.TYPE OF SPINAL DYSRAPHISM

1,2,3,4	Posterior open spinal dysraphism
5,6,7,8,9	Posterior closed with subcutaneous mass
10, 11,12,13	Posterior closed without subcutaneous mass with cutaneous stigmata
14,15,16,17,18,19,20,21	Anterior spinal dysraphism
22,23,24,25,26,27,28,29,30,31,32,33,34	Dysraphism confined to the spinal canal
35,36,37,38,39	Dysraphism involving multiple anatomical compartments
40,41	Nondysraphic masses of the spinal region

A. POSTERIOR SPINAL DYSRAPHISM

POSTERIOR OPEN- **1** Myelomeningocele

2 Myelocele

3 Hemimyelomeningocele

4 Hemimyelocele

POSTERIOR CLOSED- <u>With subcutaneous mass:</u>

5 Meningocele

6 Saccular limited dorsal myeloschisis

7 Myelocystocele

8 Posterior lipomyelocele (spinal lipoma Types IB, IIB, IIIB,)

9 Posterior lipomyelomeningocele (spinal lipoma type VB)

- <u>Without subcutaneous mass: (With cutaneous stigmata)</u>

10 Other than a dermal sinus: Nonsaccular limited dorsal

11 Myeloschisis

12 Post vertebral neurenteric cysts

13 Dorsal dermal sinus

B. ANTERIOR SPINAL DYSRAPHISM

14 Anterior myelomeningocele

15 Anterior meningocele

16 Anterior myelocele

17 Anterior myelocystocele

18 Anterior lipomyelocele (spinal lipoma type IC, IIC, IIIC)

19 Anterior lipomyelomeningocele (spinal lipoma type VA)

20 Intrasacral meningocele

21 Prevertebral neurenteric cysts

C. DYSRAPHISM CONFINED TO THE SPINAL CANAL

22 Intramedullary spinal lipoma

23 Intradural lipomas

24 Tight filum terminale (type IV D)

25 Intrasacral meningocele

26 Persistent terminal ventricle

27 Retained medullary cord

28 Split cord malformations

29 Types 1 and 2 diastematomyelia

30 Composite type diastematomyelia

31 Partial type diastematomyelia

32 Neuroenteric cysts

33 (Epi)dermoid/dermoid

34 Segmental spinal dysgenesis: type 1

D. DYSRAPHISM INVOLVING MULTIPLE ANATOMICAL COMPARTMENTS

35 Dorsal enteric fistula

36 Vertebral duplication

37 Caudal regression syndrome

38 Segmental spinal dysgenesis: type 2

39 OEIS, VACTERL, Currarino's triad associations

E. NONDYSRAPHIC MASSES OF THE SPINAL REGION

40 Sacrococcygeal teratoma

41 Sacral chordoma

II. SYMPTOMS

0-asymptomatic

1-urinary incontinence

2-single limb paresis

3-paraparesis

4-paraplegia

5-unilateral limb shortening

6-lower lumbar swelling

Sr No.	AGE(MONTHS)	GENDER	GENETIC PREDILECTION	CONSANGUINEOUS MARRIAGE	MATERNAL NUTRITIONAL STATUS	FOLIC ACID SUPPLEMENTATION	GESTATIONAL AGE AT BIRTH (WEEKS)	SYMPTOMS	TYPE OF SPINAL DYSRAPHISM	ASSOCIATED VERTEBRAL ANOMALIES	ASSOCIATED RENAL ANOMALIES	ASSOCIATED COMPLICATIONS
1	36	F	NO	NO	GOOD	YES	40	1, 3	8, 11			
2	16	M	NO	NO	GOOD	YES	34	0	13,33			
3	24	F	YES	YES	GOOD	YES	37	4	9,11		BILATERAL MODERATE HYDRON	
4	48	F	NO	NO	POOR	NO	40	1,3	24,29			
5	72	F	NO	NO	GOOD	YES	35	1	24	Dorsal butterfly vertebrae	ECTOPIC LEFT KIDNEY	
6	8	F	NO	NO	GOOD	YES	31	4	5,24,26,29			
7	60	M	YES	YES	GOOD	NO	36	0	1,11			
8	0.5	F	NO	NO	POOR	YES	38	0	1,24	CHIARI MALFORMATION I		
9	16	M	NO	NO	GOOD	YES	35	4	1,11,24,29	Lumbar butterfly vertebrae		
10	72	F	NO	YES	GOOD	YES	40	5	9,11,31	Cervical block vertebrae		
11	12	M	NO	NO	GOOD	YES	40	1	13,24,31			
12	72	M	YES	NO	GOOD	NO	38	1	13,24,31			
13	24	M	NO	NO	POOR	YES	37	0	24,29		ECTOPIC LEFT KIDNEY	
14	84	M	NO	YES	GOOD	YES	36	1,6	7,24			
15	8	F	NO	NO	GOOD	YES	38	0	8,13,24			
16	2	M	NO	NO	GOOD	YES	39	0	13,24,31			
17	96	M	NO	YES	GOOD	NO	40	0	8,13,24			
18	0.5	M	NO	NO	POOR	YES	35	6	9,24			WITH RUPTURE
19	0.5	M	NO	NO	GOOD	YES	37	6	5,24,29			EXTRASPINAL COLLECTION
20	2	M	NO	NO	GOOD	YES	32	6	8,24,30			
21	0.5	F	YES	NO	GOOD	YES	38	0	13,24,31			
22	9	F	NO	NO	GOOD	NO	41	0	11,23,24,30			
23	0.5	F	NO	YES	POOR	YES	35	6	9,24			
24	6	M	NO	NO	POOR	YES	40	1	9,24			
25	4	F	YES	NO	GOOD	YES	38	6	5,24			
26	48	F	NO	NO	GOOD	YES	41	1	7,24			
27	12	F	NO	NO	GOOD	NO	39	6	8,24	Lumbar block vertebrae with bifid spinous process		
28	0.5	M	NO	NO	GOOD	YES	36	6	1,24			
29	0.5	M	NO	NO	GOOD	YES	35	6	5			
30	72	F	NO	NO	POOR	YES	36	6	24			
31	60	M	NO	NO	GOOD	YES	38	6	7,24		BILATERAL MODERATE HYDRON	
32	48	M	NO	NO	POOR	YES	40	1,6	9,24			
33	60	F	YES	NO	GOOD	YES	34	6	23,24			
34	7	F	YES	NO	GOOD	YES	35	0	13,24,33			
35	36	F	NO	NO	GOOD	YES	40	1,6	24	CHIARI MALFORMATION II		
36	48	F	NO	NO	GOOD	YES	38	6	7,13,24			
37	96	M	NO	NO	POOR	YES	37	1,6	11,13,24,30	Dorsal bifid spinous process		
38	7	M	YES	YES	POOR	NO	34	0	37			
39	4	F	YES	NO	GOOD	YES	38	6	5,24			
40	0.5	M	NO	NO	GOOD	YES	36	6	1,24			
41	72	F	NO	NO	POOR	YES	36	6	24			
42	48	M	NO	NO	POOR	YES	40	1,6	9,24	Lumbar block vertebrae with bifid spinous process		
43	12	F	NO	NO	GOOD	NO	39	6	8,24			
44	0.5	M	NO	NO	GOOD	YES	36	6	1,24	Dorsal butterfly vertebrae		
45	96	F	NO	NO	GOOD	YES	41	1,6	7,24			
46	7	F	YES	NO	GOOD	YES	34	1	13,24,31			
47	0.5	F	YES	NO	GOOD	YES	38	0	13,24,31			
48	7	M	NO	NO	POOR	NO	33	0	11,23,24,30			
49	0.5	M	NO	NO	GOOD	YES	36	6	1,24			
50	74	F	NO	NO	GOOD	YES	38	6	7,13,24			
51	48	M	NO	NO	POOR	YES	40	1,6	9,24			
52	62	F	NO	NO	GOOD	YES	38	6	7,13,24	Lumbar block vertebrae		
53	1	M	YES	NO	GOOD	YES	36	6	1,24		ECTOPIC LEFT KIDNEY	
54	36	F	NO	YES	POOR	YES	36	6	24			
55	8	F	YES	NO	GOOD	YES	35	0	13,24,33			