11 years old male patient presented clinically with shortness of the left leg, scoliosis, pes cavus and penetrating deep ulcer in the sole of the left leg. Examination of the back revealed lumbosacral hypertrichosis.

Figure 1. Plain X ray of a case with occult spinal dysraphism showing scoliosis, spina bifida, klippel - Feil anomaly, and dilated vertebral canal
Figure 2. CT myelography showing spinal dysraphism, notice the low, tethered splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space. Notice the splitted spinous process, and the spina bifida.

Figure 3. CT myelography showing spinal dysraphism, Notice the dilated, fusiform, subarachnoid space, splitted spinal cord and the spina bifida. An anterior vertebral segmentation defect is seen resulting in almost a bifid vertebral body.

Figure 4. CT myelography showing spinal dysraphism, notice the fibrous band that separates the splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space. Each splitted hemicord is enclosed in a separate arachnoid sac (dural tube). This is frequently termed Type II diastematomyelia. The two hemicord are not equal in size.
**DIAGNOSIS:**

**DISCUSSION:**

The spinal cord is formed by invagination of a tube of ectoderm, around which the mesoderm and scleroderm close to
form the meninges and vertebral column. This process when interrupted before completion, results in malformations ranging from minor radiological abnormalities to those incompatible with life.

The general term for these malformations is dysraphism. In order of severity, the grades of dysraphism are:

1- Overt spinal dysraphism

2- Occult spinal dysraphism

Congenital spinal anomalies have been classified into three broad categories - 1) Open spinal dysraphism 2) Occult spinal dysraphism and 3) Caudal spinal anomalies. Occult spinal dysraphism refers to a heterogenous group of disorders in which the neural tissue lies deep to an intact skin cover. Open dysraphism is characterized by exposed neural tissue and the condition is clinically evident at birth. In caudal spinal anomalies, the clinical picture is dominated by anorectal, urogenital and lower limb anomalies [1]. Occult spinal dysraphism, tend to be overlooked at birth. The manifestations become apparent later in life due to either cutaneous stigmata or slowly progressing neurological signs [2]. Imaging is essential to diagnose and characterize these lesions.

- Embryology

By the 17th post-ovulation day, the notochord forms in the midline of the embryo. The ectoderm overlying the notochord is induced to form the neuroectoderm. The neuroectoderm folds and dissociates from the superficial ectoderm in a process termed primary neurulation. This process explains the formation of most of the spinal cord. The distal conus and the filum terminale develop as a result of canalization and retrogressive differentiation or secondary neurulation.

**OVERT SPINAL DYSRAPHISM**

- **Simple Spina Bifida**

It is a bony defect, without herniation of meninges or nervous tissue. It occurs most commonly at the lumbosacral and first cervical levels. It is frequently asymptomatic incidental finding, but in appropriate clinical picture, should alert to the possibility of an underlying malformation of the neuraxis.

- **Meningocele and meningomyelocele**

This refers to protrusion of meninges and nervous tissue, it occurs most frequently near the foramen magnum, and in the lumbosacral region, the thoracolumbar junction is another common site. A posterior defect and widening of the spinal canal are common associated abnormalities.

Two specific types of meningoceles without a posterior defect may be encountered:

- **Anterior sacral meningocele:**

It commonly occurs in the dorsal spine and protrudes through an intervertebral foramen. Lateral meningoceles are frequently encountered in patients with neurofibromatosis.
OCCULT SPINAL DYSRAPHISM

This term is applied to cases in which intraspinal anomalies are more striking than manifestation of impaired closure.

The most common anatomical abnormality of the spinal cord in occult spinal dysraphism is diastematomyelia which is a sagittal division or pseudoduplication of a segment of the spinal cord, diastematomyelia most commonly occurs in the lower dorsal and upper lumbar region. In many cases, the two cords are contained within a single dural tube, but in others, each of the two cords has its own dural sheath and the two are separated by a bony or fibrocartilagenous septum. Low cord termination, below L 2 posterior tethering of the cord and thick filum terminate are common associated anomalies.

Figure 7. Anterior meningocele

Figure 8. The back of two patients with occult spinal dysraphism, notice the hairy tuft overlying subcutaneous lipomas, these lipomas often extend through a spinal bifida defect to and usually through the dura, and might exert a considerable mechanical pressure against nerve roots and spinal cord
The cord is commonly transfixed at the point of diastases by the osseous or fibrocartilagenous septum which is attached anteriorly to one or more vertebral bodies and posteriorly to the dura. It passes through the spinal cord and fixes it at low anatomical position. Arnold-chiari malformation may be encountered as the normal ascend of the spinal cord will be arrested. Hydrocephalus is occasionally associated with spinal dysraphism. (Click for a short case)

Figure 9. Chiari malformation in two cases with occult spinal dysraphism

Congenital anomalies of the vertebral column are frequently encountered in spinal dysraphism. Widening of the vertebral canal is a common finding and might extend for as many as six vertebral segments. The interpedicular distance is increased in a fusiform manner over these segments. Numerous developments abnormalities of the vertebral bodies and arches (such as Klippel feil syndrome) may be present in association with the widening of the vertebral canal.

In general, defects in primary or secondary neurulation constitute the primary pathogenesis of spinal dysraphism and explain most of the pathology demonstrated in this disorder.

- **Pathology due to defects of primary and secondary neurulation**
  - **Lipomas**

A congenital tumour, commonly dermoid or lipoma might be present and might fill the lower spinal canal The cord or the film terminate may pass directly into the tumour which might be intra-dural or extra dura. Lipoma is the most common type of occult spinal dysraphism. Spinal lipomas are more common in females. In infancy, the presentation is usually as a mass in the back. Later in life, pain, motor deficits, urinary incontinence and foot deformities are common presenting features [1, 3].

A classification based strictly on the embryology divides lipomas into posterior (retromedullary) epidural lipoma, transitional lipoma and terminal lipoma [4]. Both posterior (retromedullary) epidural and transitional lipomas have an intraspinal component which blends with the cord and a fibro-fatty stalk that fuses with the subcutaneous fat. In posterior (retromedullary) epidural lipomas, however, the cord distal to the attachment of lipoma is entirely normal. These are considered as defects of primary neurulation and constitute less than 10% of all spinal lipomas. If the superficial ectoderm separates from the neuroectoderm before the neural tube has completely closed (premature disjunction), then the mesoderm comes into contact with interior of the neural tube. This would prevent complete closure of the neural tube. The mesoderm in contact with the interior of the neural tube is induced to form fat [1].
In transitional lipomas, the distal cord and conus terminate in the lipoma and there is no normal cord caudal to the lipoma. Since the distal conus is derived from a process of secondary rather than primary neurulation, defects of both processes have to be postulated to explain the condition - hence the term ‘transitional’. The common lumbosacral lipomyelomeningocele is a transitional lipoma. A defective process of canalization and retrogressive differentiation alone results in a terminal lipoma represented by lipomas of the filum terminale.

Intradural or spinal cord lipomas are, probably, a rare form of posterior (retromedullary) epidural lipoma, comprising approximately 4% of all spinal lipomas and 1% of all spinal cord tumours. Spina bifida is usually absent or small, if present. These lipomas are most commonly seen in the cervico-thoracic region.

- **Tight filum syndrome**

  Tight filum syndrome represents a failure of retrogressive differentiation and is part of the same pathogenetic continuum as filar lipoma. The filum should measure more than 2 mm in thickness there should be no other cause for tethering. Conus medullaris is elongated with no sharp transition between the conus and filum. Almost all patients have a spina bifida. The tethered cord may terminate in a lipoma [1, 3].

- **Terminal syringohydromyelia**

  Expansion and cephalic extension of the terminal ventricle within the distal conus can occur involving the distal one thirds of the cord. This can be seen in upto 30% of patients with occult spinal dysraphism or can occur alone [5]. On rare occasion diastematomyelia may occur in the cervical region and may give a clinical picture resembling that of congenital syringomyelia.

- **Dorsal Dermal sinus**

  During the process of primary neurulation, the neuroectoderm may fail to separate from the superficial ectoderm at one point (non-disjunction), as development proceeds this adhesion gets drawn out into an epithelial lined tube connecting the spinal cord to the skin of the back. This epithelial track is termed dorsal dermal sinus [1]. The lumbosacral region is the commonest location. The sinus tract may extend superficial to the dura, but in more than 50% of cases it extends into the spinal canal. In 60% of cases the tract incorporates or ends in an epidermoid or dermoid [1]. CSF leak through the sinus tract, is rare, and can lead to meningitis [1].

Almost 50% of spinal epidermoids and dermoids occur without an associated spina bifida or dermal sinus. An inadvertent inclusion of cutaneous ectoderm during neural tube closure is thought to result in these isolated lesions.

- **Split notochord and related disorders**

  These conditions result from a persistent adhesion between the entoderm and the ectoderm in the midline which interferes with the normal formation of the notochord. The notochord is bisected and thereby inducing the formation of two hemicords.

  - **Diastematomyelia**

    More than 80% of cases are seen in females. Presentation is typically with orthopaedic deformities like clubfoot. Segmentation anomalies of the vertebrae like hemivertebrae, butterfly vertebrae and block vertebrae are seen in most patients. The spinal cord is split sagitally into two halves by a fibrous or osteocartilaginous septum. The conus lies low and is tethered. Two distinct types of diastematomyelia have been described. In slightly less than 50% cases, each hemicord lies within its own dural tube and in nearly all these cases, there is a bony septum. In the rest, the two hemicords lie within a single dural tube. In these patients there will not be a bony spur [1, 3].

    - **Neurenteric cyst**

      These are cysts, which have a definite connection with the spinal cord and vertebra and are lined with alimentary tract mucosa. The cyst may be extraspinal, in the posterior mediastinum, with an intraspinal component that invaginates the cord. An anterior or posterior spina bifida is usually associated. The cyst may also be entirely intraspinal without any spina bifida [1, 7].

**CLINICAL PICTURE**
Lumbosacral spinal dysraphism has been held responsible for a variety of disturbances which are occasionally familiar. The principle symptoms are impairment of sphincter control; deformities of the feet; wasting of the muscles below the knees, with impairment of the ankle-jerks; dissociated sensory loss of a syringomyelic character over one or both legs and trophic disturbance of the feet such as delayed healing of wounds; chronic ulceration and gangrene.

**RADIOLOGICAL EVALUATION**

Plain X-ray commonly demonstrates spina bifida, widening of interpedicular distance, scoliosis, and bony spurs.

Pantopaque myelography was used in the past to confirm occult cases of spinal dysraphism. The contrast material was so dense that many of the subtle features of these anomalies are not demonstrated until the time of surgical exploration. MRI and metrizamide myelography, especially in combination with CT scanning has offered significantly better visualization of the often multiple features of spinal dysraphism.

The pathognomonic CT or MRI findings in diastematomyelia is two hemicords of abnormally small and often unequal size within the spinal canal. Because diastematomyelia is usually associated with a large subarachnoid space, the abnormal hemicords can be demonstrated by plain CT as well as after intrathecal enhancement. The rostrocaudal extent of diastematomyelia can be determined by inspecting contiguous CT slices, by generating sagittal and coronal reconstructions or much better by MRI. The two hemicords are separated by either bony spares or fibrocollagenous bands.

![Intraoperative images (A,B) showing spinal dysraphism, notice the bony spur that separate the splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space (B)](image)

**Figure 10.** Intraoperative images (A,B) showing spinal dysraphism, notice the bony spur that separate the splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space (B)
Figure 11. CT myelography (A,B) showing spinal dysraphism, notice the bony spur that separate the splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space (B)

The more sensitive density discrimination afforded by CT or MRI has resulted in the identification of both extraspinal and intraspinal lipomas and in their clear demarcation from an associated tethered cord. Lipomas are characteristically hyperintense on precontrast MRI T1 images.

Figure 12. MRI T1 images showing diastematomyelia. Notice the hemicords and fusiform dilatation of the lumbar canal.

Lipomas are the most frequent tumours in spinal dysraphism. Although, they are occasionally limited to the intradural compartment, however these lesions are commonly present at birth as subcutaneous collection of fat in the midline over the lumbosacral area and extend through a spinal bifida defect to and usually through the dura.
The intraspinal portion of the lipoma might exert a considerable mechanical pressure against nerve roots and spinal cord. Pre-operative demonstration of these lipomas is carried out by CT scan or MRI.

Although ossified spurs can be detected by plain radiographs, but they are unmistakable in CT scan cross sectional analysis. Fibrocartilagenous soft tissue spurs can only be detected by CT scan or much better by MRI. CT scan or MRI are also the best diagnostic modality to visualize a thick film terminate tethering the cord from below. If this goes undetected, then even though the diastematomyelic spur is removed satisfactorily, the cord may remain tethered from below.

Figure 13. CT myelography (A,B) and myelography (C) showing spinal dysraphism, notice the bony spur (A) and the fibrous band (C) that separate the splitted spinal cord. Also notice the dilated, fusiform, subarachnoid space (C), notice the shadow of the splitted spinal cord in the myelographic image (C)
Figure 14. A case of spinal dysraphism with a large lipoma expanding the spinal canal, the lipoma is hyperintense on precontrast MRI T1 image (B) and a low tethered cord.

Figure 15. A case of diastematomyelia. MRI T1 precontrast (A,B) and MRI T2 images (C,D) showing low tethered cord and epidural lipomas.
Figure 16. Klippel-Feil anomaly with diastematomyelia. A, Midsagittal T1-weighted MR scan reveals partial or complete congenital nonsegmentation of the second through fifth cervical vertebrae. The C1 ring is abnormally positioned such that the anterior atlas arch (dot) lies superior to the odontoid process (0), which is abnormally separated from the basion (arrow). A degree of basiocciput hypoplasia is present such that the clivus appears shortened, the basion located at the level of the pontomedullary junction. A Chiari I malformation is present with the tonsils displaced more than 5 mm below the plane of the foramen magnum (dotted line). B, Axial T1-weighted MR scan additionally reveals the presence of cervical diastematomyelia.

Table 1. Pathological findings in occult spinal dysraphism

<table>
<thead>
<tr>
<th>Anatomical structure</th>
<th>Pathological findings</th>
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<tr>
<td>Bony vertebrae</td>
<td>Widening of the vertebral canal is a common finding and might extend for as many as six vertebral segments. The interpedicular distance is increased in a fusiform manner over these segments. Numerous developments abnormalities of the vertebral bodies and arches (such as Klippel feil syndrome) may be present in association with the widening of the vertebral canal.</td>
</tr>
<tr>
<td>Subarachnoid spaces</td>
<td>The subarachnoid spaces are enlarged and fusiform in shape</td>
</tr>
<tr>
<td>The spinal cord</td>
<td>The cardinal pathological finding in diastematomyelia is two hemicords of abnormally small and often unequal size within the spinal canal. Because diastematomyelia is usually associated with a large subarachnoid space, the abnormal hemicords can be demonstrated by plain CT as well as after intrathecal enhancement. The rostrocaudal extent of diastematomyelia can be determined by inspecting contiguous CT slices or by generating sagittal and coronal reconstructions or much better by MRI. The two hemicords are separated by either bony spares or fibrocollagenous bands. The cord is commonly transfixed at the point of diastases by the osseous or fibrocartilagenous septum which is attached anteriorly to one or more vertebral bodies and posteriorly to the dura. It passes through the spinal cord and fixes it at low anatomical position. Arnold-chiari malformation may be encountered as the normal ascend of the spinal cord will be arrested. The spinal cord, apart from its being splitted into two hemicords of unequal size, is characteristically demonstrated in a lower than normal anatomical position (often in the lumbar vertebral canal) and tethered from below.</td>
</tr>
<tr>
<td>Lipoma</td>
<td>Lipomas are the most frequent tumours in spinal dysraphism. Although, they are occasionally limited to the intradural compartment, however these lesions are commonly present at birth as subcutaneous collection of fat in the midline over the lumbosacral area and extend through a spinal bifida defect to and usually through the dura.</td>
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SUMMARY

Spinal dysraphism, or neural tube defect (NTD), is a broad term encompassing a heterogeneous group of congenital spinal anomalies, which result from defective closure of the neural tube early in fetal life and anomalous development of the caudal cell mass.

Some forms of spinal dysraphism can cause progressive neurologic deterioration. The anatomic features common to the entire group is an anomaly in the midline structures of the back, especially the absence of some of the neural arches, and defects of the skin, filum terminale, nerves, and spinal cord.

Spinal dysraphism can be classified as closed forms or open forms, which include myelocele, meningocele, and myelomeningocele. These open forms are often associated with hydrocephalus and Arnold-chiari malformation type II and may be classified as spina bifida aperta. Von Recklinghausen (1886) gave a detailed account of spina bifida cystica (aperta). Spina bifida is described in the medieval literature, although the condition was recognized even earlier. Indeed, the association of foot deformities with lumbar or lumbosacral hypertrichosis may be the origin of the mythological figure of the satyr.

The closed form of spina bifida is termed spina bifida occulta. 5-10% of the general population may have bony spina...
bifida occulta with intact overlying skin. Most of these cases are found incidentally.

Open neural tube defects (ONTDs) represents a serious congenital anomaly. If the neural tube fails to fuse at the skull, the result may be that of anencephalus or encephalocoele. If the tube fails to fuse along the spine, the resulting defect is an open neural tube defect such as meningomyelocele. Infants with neural tube defects frequently have additional serious neurologic, musculoskeletal, genitourinary, and bowel anomalies.

Spina bifida occulta is characterized by variable absence of several neural arches and various cutaneous abnormalities, such as lipoma, hemangioma, cutis aplasia, dermal sinus, or hairy patch, and it is often associated with a low-lying conus and other spinal cord anomalies. Whenever the conus lies below the L2-3 interspace in an infant, cord tethering should be considered. The term tethered cord implies that the cord may be attached to vertebral column or subcutaneous tissues by a thickened filum terminale, fibrous band, dermal sinus tract, diastematomyelia, or a lipoma (lipomyelomeningocele). Patients with spina bifida occulta may present with scoliosis in later years.

Approximately 95% of couples that have a fetus affected with ONTD have a negative family history. Most ONTDs are caused by multifactorial inheritance, including genetic and environmental factors.

- **Addendum**
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