REVIEW OF CONGENITAL SPINAL AND SPINAL CORD MALFORMATIONS

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Introduction
Spinal dysraphism

Congenital malformations of spine and spinal cord
Spinal dysraphism

Heterogeneous group of anomalies resulting from *incomplete midline closure of osseous, mesenchymal and nervous tissue*
Spinal dysraphism

Diagnosed:

- soon after birth
- late, in childhood or in adulthood

Absence of clinical manifestations
Spinal dysraphism

MRI

Modality of choice for diagnostic

Superior soft tissue characterisation and multiparametric imaging capabilities
Embryology
## Embryology

### Development of spinal cord

**Early embryogenesis**

*2 – 6 wk of gestation*

<table>
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<tr>
<th>3 main stages</th>
<th>Gastrulation</th>
<th>Primary neurulation</th>
<th>Secondary neurulation</th>
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Primary neurulation

- Brain
- Uppermost 9/10 of spinal cord

Secondary neurulation

- Tip of conus medullaris
- Filum terminale
Primary neurulation

The zipper-like model
Secondary neurulation

CAUDAL CELL MASS

CANALIZATION

MERGE

1 RY NEURAL TUBE

2 RY NEURAL TUBE

RETROGRESSIVE DIFFERENTIATION

CONUS MEDULLARIS

V.T + F.T
Malformation secondary to derrangement of any of these steps is called spinal dysraphism.
Classification

Open × closed
overlying skin covering
Spinal Dysraphism (Classification)

**Open type**

Overlying skin covering is **absent**

+ Abnormal neural tissue directly exposed to the external environment
Spinal Dysraphism (Classification)

Closed type (occult)

The neural elements have a skin covering

Presence or absence of associated subcutaneous mass:

- With
- Without
Spinal Dysraphism (Classification)

Presence or absence of overlying skin covering

**Closed type**

The neural element have a skin covering

Presence or absence of associated subcutaneous mass:

*With* - -  

*Without* - -
Spinal Dysraphism (Classification)

Presence or absence of overlying skin covering

- Open type: Overlying skin covering is absent
- Closed type: Abnormal neural tissue directly exposed to the external environment

Presence or absence of associated subcutaneous mass:
- With
- Without
<table>
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<th>Clinic-radiological classification</th>
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<td>* Myelomeningocele and myelocele</td>
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<td><strong>CSDS</strong></td>
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<td><strong>With subcutaneous mass</strong></td>
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<td>* Lipomas with dural defect:</td>
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<tr>
<td>lipomyelocele and lipomyelomeningocele</td>
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<tr>
<td>* Meningocele</td>
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<td>* Terminal myelocystocele</td>
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<td><strong>Without subcutaneous mass:</strong></td>
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<td>* Intradural lipoma</td>
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<td>* Tight filum terminale</td>
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<td>* Dermal sinus</td>
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<td>* Persistant terminal ventricle</td>
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<td>* Disorders of midline notochordal integration:</td>
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<td>- Neurenteric cyst</td>
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<td>- Caudal agenesis defect</td>
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Classification
PLACODE

Undifferentiated segment of the spinal cord who’s development is arrested at the neural plate stage
Open spinal dysraphisms (OSDs)
Dorsal herniation of all or part of the contents of the spine. Both the neural placode and meningeal lining protrude through the bony and cutaneous defect in the midline.
OSDs

Dorsal herniation of all or part of the contents of the spine. Both the neural placode and meningeal lining protrude through the bony and cutaneous defect in the midline.

Myelomeningocele
OSDs

Dorsal herniation of all or part of the contents of the spine. Both the *neural placode* and *meningeal lining* protrude through the *bony and cutaneous defect* in the midline.

**Myelomeningocele**

**Myelocele**

All OSDs are anomalies of 1ry neurulation.
The placode is flush with the skin surface
placode at same level as surrounding skin with very large fat interruption
The placode lies above the skin surface due to cystic dilatation of the subarachnoid spaces.
OSDs

Both the neural placode and meningeal lining protrude through the bony and cutaneous defect in the midline.
OSDs (MMC) are typically associated with Chiari II malformation

- CSF leakage
- Failure to expand the rhomboencephalic vesicle
- Small posterior cranial fossa
- Chiari II malformation
  *Various manifestations*
The hallmark of the Chiari II malformation

A small, crowded posterior fossa with downward herniation of the vermis
## OSDs

<table>
<thead>
<tr>
<th>Clinicaly</th>
<th>The neonate presents with midline reddish exposed neural placode</th>
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<tbody>
<tr>
<td>Imaging</td>
<td>Not always performed</td>
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<tr>
<td></td>
<td>Immediate surgical repair is usually done</td>
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<td></td>
<td>Protruding neural placode extends beyond the skin surface</td>
</tr>
<tr>
<td></td>
<td>Enlargement of the adjacent subarachnoid space</td>
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<td>Associated hydrocephalus</td>
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<tr>
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<td>Chiari II malformation</td>
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<td></td>
<td>Involve cerebellum, brain stem, skull base, spine and spinal column</td>
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**Diagnosis** of neural tube defects in fetus as early as first trimester is possible: Sonography – MRI - faster MR sequences -

**Fetal surgery** for MMC in second trimester preserves neurologic function, reverses the changes of Chiari II malformation and reduces the need for postnatal ventriculoperitoneal shunt.
OSDs

Hemimyelomeningocele and hemimyelocele

Defective gastrulation and primary neurulation

One of the hemicords shows defective neurulation

Diastematomyelia is associated with

One of the hemicords exhibits a ……

Myelomeningocele

Hérmimyelomeningocele

Myelocele

Hemimyelocele
Closed spinal dysraphisms (CSDs)
1. Lipomyelocele
2. Lipomyelomeningocele
3. Meningocele
4. Terminal myelocele
1. Lipomyelocele
2. Lipomyelomeningocele
3. Meningocele
4. Terminal myelocele

Malformation showing a masse
1. Lipomyelocele
2. Lipomyelomeningocele
3. Meningocele
4. Terminal myelomeningocele
CSDs

Lipomas with dural defect

Mechanism: Defective 1 ry neurulation

Premature focal disjunction of cutaneous ectoderm and neuroectoderm allowing mesenchyme to enter the neural tube.

Later, forms the lipomatous tissue

*Unknown reasons*
| **Clinically** | Presence of subcutaneous fatty mass lesion above the intergluteal line which may extend to buttocks |
| **MRI** | * **High intensity** fat on the dorsal aspect of the placode which continuous with the adjacent subcutaneous fat |
| **T1, T2, T1 W FAT SAT** | Suppression of fat signal |
Lipomyelocele
lipomyelomeningocele

dural defect

Presence of dural defect through which the lipoma may extend from the spinal cord to the subcutaneous tissue
Differentiated based on the position of neural placode-lipoma interface

- Lies within or at the edge of the spinal canal
- Lies outside the spinal canal
CSDs

Lipomyelocele

Lipoma creeps through to connect with the spinal canal
CSDs

Lipomyelocele

The placode-lipoma interface lies within the spinal canal
Lipomas with dural defect

Lipomyelomeningocele

The placode lipoma interface lies outside the spinal canal
Associated meningeal herniation
## CSDs

**Lipomas with dural defect**

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Differentiated based on **the position of neural placode-lipoma interface**

- Lies within or at the edge of the spinal canal
- Expansion of subarachnoid space anterior to the cord pushing the neural placode-lipoma interface posteriorly to lie outside the boundaries of spinal canal

- Lies outside the spinal canal
CSDs

Meningocele

Herniation of CSF filled sac lined by dura and arachnoid mater

Exact embryogenesis: unknown

Ballooning of meninges due to CSF pulsation
Meningocele

Spinal cord should not be seen within the meningocele

May be seen tethered to the neck

May contain nerve roots and or filum terminale which usually appear hypertrophied
Defective secondary neurulation
Affect the CSF flow dynamics
Terminal myelocystocele

Involves herniation of dilated terminal central canal forming terminal syringohydromyelia (syringocele) through a posterior vertebral defect into an expanded CSF filled dural sheath (meningocele).

The inner terminal syrinx
Communicates with the central canal of the spinal cord

The outer meningocele
Is continuous with the spinal subarachnoid space

Syringocele and meningocele usually don't communicate with each other.
Closed spinal dysraphisms without a mass
Simple dysraphic states

- Intradural lipoma
- Filar lipoma
- Tight filum terminale
- Dermal sinus
- Persistent terminal ventricle

Primary and secondary neurulation

The most common type of spinal dysraphism seen in old children
Simple dysraphic states

- Intradural lipoma
- Filar lipoma
- Tight filum terminale
- Dermal sinus
- Persistent terminal ventricle

Primary and secondary neurulation

The most common type of spinal dysraphism seen in old children
Simple dysraphic states

**Intradural lipoma**

Midline lipoma located in the groove of unopposed neural placode in its dorsal surface within an intact dural sac

**Differentiate from lipomyelocele and lipomyelomeningocele:**

**Intact dura**

Lumbosacral region

Associated with tethered cord syndrome
Simple dysraphic states

Intradural lipoma

MRI: Lipomas follow the signal intensity of subcutaneous fat on all sequences.
Simple dysraphic states

Filar lipoma

Secondary neurulation

Fibrolipomatous thickening of the filum terminale

T1 and T2 weighted images within a thickened filum terminale
Simple dysraphic states

Filar lipoma

Secondary neurulation
Fibrolipomatous thickening of the filum terminale

T1 and T2 weighted images within a thickened filum terminale
Simple dysraphic states

*Filar lipoma*

Normal variant unless it is associated with tethered *cord syndrome*
Simple dysraphic states

Tight filum terminale

The defect occurs in retrogressive differentiation during secondary neurulation

* Shortening and hypertrophy of filum terminale which cause tethering of cord
Simple dysraphic states

**Tight filum terminale**

The defect occurs in retrogressive differentiation during secondary neurulation

* Thick filum terminale (thickness measuring > 2 mm)
  - Low lying conus medullaris – below L2 vertebral body

*Isolated cases are rare*
CSDs
Without --

**Simple dysraphic states**

*Dermal sinus*

Focal incomplete disjunction between neuroectoderm and cutaneous ectoderm

Epithelial lined fistulous communication between CNS or its meningeal covering and skin
Simple dysraphic states

Dermal sinus

these are T2 and T1 sagittal WI of lumbosacral spine showing a T2 hypointense tract extending from the posterior skin surface to the spinal canal.
CNS infection is a common complication because of fistulous communication. This cases require early surgical repair.

There is associated tethered cord. Contrast enhancement.
CSDs

Without - -

Simple dysraphic states

**Persistent terminal ventricle**
Small, ependyma lined cavity within conus medullaris

Secondary neurulation
Incomplete regression of terminal ventricle
CSDs

Simple dysraphic states

Persistent terminal ventricle
Small, ependyma lined cavity within conus medullaris

Above filum terminal
Δ hydromyelia

Lack of enhancement
Δ intramedullary tumors
Gastrulation

Abnormalities affects the spinal cord and various other structures which are derived from notochord.

Subcutaneous masses are absent
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| **Segmental spinal dysgenesis** |
Neurenteric cyst
Localized form of dorsal enteric fistula
Seen anterior to spinal cord with adjacent vertebral anomalies
Typically seen in extramedullary intradural compartment of cervicothoracic spine
*May be seen in other locations*
Neurenteric cyst

MRI:
Iso- to hyperintense to CSF on both T1 and T2 weighted images

Due to high protein content

Absent contrast enhancement
Diastematomyelia

*The most common form*

- Defective midline integration
  - Two notochordal processes
  - Development of primitive streak tissue
  - Type 1
  - Type 2

Complex dysraphic states

CSDs

Without --
Complex dysraphic states

Diastematomyelia

Type 1

Primitive streak

Develops into bone or cartilage

2 hemicords in different dural sacs separated by an osteocartilaginous septum

Type 2

Primitive streak

Resorbed

Forms a fibrous septum with the hemicords lying within the same dural sac

CSDs
Without - -
Diastematomyelia I. Axial T2 weighted scan of lumbar spine showing two hemicords. Intervening bony septum.
CSDs
Without --

Complex dysraphic states

Diastematomyelia
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**Type 1 CA**
- **Affected**: Both caudal cell mass and notocord
- High level (D12)
- Abnormal termination of conus medullaris
- Accompanying varying degree of vertebral aplasia (L5 > S2)

**Type 2 CA**
- Abnormal development of only caudal cell mass
- Unaffected true notocord formation
- Absence of the most caudal part of conus medullaris
Complex dysraphic states

Caudal agenesis

T1 and T2 W Sagittal images showing type I caudal agenesis
There is abnormal termination of conus medullaris with non-development of distal sacral vertebrae.
Group of diverse conditions

Systematic approach and correlation with neuroradiological, clinical and developmental data helps in making diagnosis

Variable imaging appearance
bibliography

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