Spine and Spinal Cord Malformations

Embryological classification of spinal dysraphisms

Gastrulation (2-3 W)
Primary neurulation (3-4 W)
Secondary neurulation (4-6 W)

SEGMENTAL NOTOCHORDAL FORMATION
Caudal Regression Syndrome
Segmental Spinal Dysgenesis

Primary neurulation
ABSENT NEURULATION
Myelomingingocele
Myelocele
Hemimeilocele

SECONDARY NEURULATION
Lipomyelomingingocele
Lipomyelocele

MANDIBLE DEVIATION
Split Notochord Syndrome
Diastematomyelia

LATE DISJUNCTION
Dermal sinus
Nonterminal Myelocystocele
Meningocele

RETROGRESSIVE DIFFERENTIATION
Filum Terminale Lipoma
Tight Filum Terminale

Workaday classification of spinal dysraphisms

Which of the following malformations can be ruled out based on the external features?

1. Lipomyeloschisis
2. Lipomyelomeningocele
3. Myelomeningocele
4. Myelocystocele
5. Meningocele

A myelomeningocele is an open spinal dysraphism (i.e., abnormal neural tissue is exposed)
The initial diagnostic evaluation of patients with suspected spinal dysraphisms (SD) is a **clinical, not imaging, issue** that radiologists should personally address.

**Open Spinal Dysraphisms**
- Dorsal herniation of all or part of the contents of the spine
- Undifferentiated neural tissue (placode) is constantly exposed

**Myelomeningocele**

*Myelocele, Hemimyelocele, Hemimyelomeningocele*

All **OSDs** are anomalies of primary neurulation.

**Myelomeningocele**

The placode lies above the skin surface due to cystic dilatation of the subarachnoid spaces.

**Myelocele (syn. Myeloschisis)**

The placode is flush with the skin surface.

Open spinal dysraphisms (i.e., myelomeningoceles) are typically associated with the **Chiari II malformation**

- CSF leakage
- Failure to expand the rhombencephalic vesicle
- Small posterior cranial fossa w/hermations

McLone and Knepper, 1989
Fetal head biometry assessed by fetal magnetic resonance imaging following in utero myelomeningocele repair. Fetal Diagn Ther 2007; 22:1-6.

**Workaday classification of spinal dysraphisms**

<table>
<thead>
<tr>
<th>Open SD</th>
<th>Closed SD</th>
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<tbody>
<tr>
<td>WITH SUBCUTANEOUS MASS</td>
<td>WITHOUT SUBCUTANEOUS MASS</td>
</tr>
<tr>
<td>Myelomeningocele</td>
<td>Myelocele</td>
</tr>
<tr>
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**Lipomyelolele**
(syn. Lipomyeloschisis)

The placode-lipoma interface lies within the spinal canal.
Lipomyelomeningocele

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

Lipomyelocele

Typical *Lumbo-Sacral* CSD with tumefaction

They differ from one another in the position of the placode-lipoma interface
Terminal myelocystocele

- Herniation of a hugely dilated terminal ventricle ("syringocele") into a meningocele
- Possible association with the OEIS complex

Myelocystocele

Closed spinal dysraphisms with tumefaction

Lipomyelocele
Lipomyelomeningocele
Myelocystocele
Meningocele

Closed spinal dysraphisms with tumefaction

Cord-lipoma junction within the spinal canal
Cord-lipoma junction outside the spinal canal
"Cyst within a cyst"

Terminal myelocystocele

Herniation of a hugely dilated terminal ventricle ("syringocele") into a meningocele
Possible association with the OEIS complex

Lipomyelocele
Lipomyelomeningocele
Myelocystocele
Meningocele

Open SD
Closed SD

Myelomeningocele
Myelocele
Hemimyelo(meningo)cele

Myelomeningocele
Myelocele
Hemimyelo(meningo)cele

Lipomyelocele
Lipomyelomeningocele
Myelocystocele
Meningocele

Workaday classification of spinal dysraphisms

Open SD
Closed SD

WITH SUBCUTANEOUS MASS

WITHOUT SUBCUTANEOUS MASS
Diastematomyelia

[Split cord malformations]

Segmental incomplete duplication of the spinal cord in which each hemicord has a single set of anterior and posterior nerve horns and roots

Diastematomyelia Type 1

The two hemicords are contained within two separate dural tubes. Intervening septum ("spur") may be osseous or cartilaginous, symmetric or asymmetric, complete or incomplete. Each hemicord usually has a single set of anterior and posterior nerve roots. "Paramedian" nerve roots may originate medially and connect to the spur.

Diastematomyelia Type 2

The two hemicords are housed within a single dural sac.
Hemimyelo(meningo)cele

Splitting of the spinal cord (i.e., diastematomyelia), in which one hemicord fails to neurulate

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- Meningomyelocele
- Lipomyelocele
- Lipomyelomeningocele
- Myelocystocele
- Meningocele

'SPLIT' CORD
Diastematomyelia

Tethered Cord Syndrome

- Not a malformation, but rather a clinical constellation
- Caused by several CSDs (i.e., tight filum, lipomas, diastematomyelia) as well as by scarring following OSD surgery
- Association of neurogenic bladder, sphincter incontinence, gait disturbances, lower limb deformities, scoliosis
- The conus apex is low (i.e., below L2-3)
- Relieved by surgical detethering

Isolated Filar lipoma in CRS Type II

Filar lipoma

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Workaday classification of spinal dysraphisms

Open SD
- Myelomeningocele
- Myelocle
- Hemimyelo(meningo)cle
- Lipomyelocele
- Lipomyelomeningocele
- Myelocystocele
- Meningocele

Closed SD
- WITH SUBCUTANEOUS MASS
- "LOW-LYING" CORD
  - Filar & Intradural Lipomas
  - Tight Filum Terminale
  - Type 2 Caudal Regression Sy
- WITHOUT SUBCUTANEOUS MASS
- "SPLIT" CORD
  - Diastematomyelia

Caudal Regression [Agenesis] Syndrome
Wide array of anomalies including, in various combinations:
- thoraco-lumbo-sacral anomalies
- genital malformations
- renal dysplasia
- pulmonary hypoplasia
- anorectal atresia

courtesy R. Silva Carvalho, S. Paulo Brasil
Caudal Regression Syndrome type 1

- Variable degree of spinal cord hypoplasia among individuals
- "Fixed" neurological deficiency correlated with degree of spinal cord hypoplasia
- "double bundle" shape
- "ghost conus" sign

Caudal Regression Syndrome TYPE 2

- Lesser degree of vertebral agenesis (below S2)
- Low-lying spinal cord terminus, usually tethered to filar or intradural lipoma, tight filum, or meningocele

Segmental spinal dysgenesis

- Congenital paraplegia
- Hypoplastic lower limbs w/ equinacavovarus deformity
- Focal aplasia of the spine and spinal cord with disconnection ("cut in two")
- Spinal cord present both above and below the dysgenesis

Tortori-Donati P & Rossi A, AJNR 1999

Workaday classification of spinal dysraphisms

Open SD
- Myelomeningocele
- Hemimyelo(mentino)xarel
- Lipomyelocele
- Lipomyelo(myelomeningo)cele
- Myelorachdystele
- Meningocele

Closed SD
- Myelomeningocele
- Myelocele
- Hemimyelocele
- Lipomyelocele
- Lipomyelo(myelomeningo)cele
- Myelorachdysplasia
- Meningocele

"LOW LYING" CORD
- Filar & Intradural Lipomas
- Tight Filum Terminale
- Type 1 Caudal Regression Sy

"SPLIT" CORD
- Diastematomyelia

"INTERUPTED" CORD
- Type 1 Caudal Regression Sy
- Segmental Spinal Dysgenesis

Heavy T2-weighting, sub-mm slice thickness

T2 DRIVE
- Philips

CISS
- Siemens

FIESTA
- GE
Atretic meningocele

Bilateral abortive nonterminal hemi-myelocystocele

Les classifications ne sont que le reflet de notre ignorance

Classifications are only a reflection of our ignorance

Pierre Lasjaunias (1948-2008)