Congenital Spine Anomalies

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OVERVIEW

- Spinal cord development in fetus
- Spinal dysraphisms, classification
  - Open spinal dysraphisms
  - Closed spinal dysraphisms
    - with subcutaneous mass
    - without subcutaneous mass
      - simple
      - complex
LEARNING OBJECTIVES

- Describe the algorithmic approach to spinal dysraphism
- Identify the
  - Neural placode
  - Lipoma-placode interface
  - Lipoma
  - Expanded subarachnoid space
  - Hemicords
  ...

LEARNING OBJECTIVES

- Fluid-filled structures
  - Expanded subarachnoid space (meningocele)
  - Hydromyelia
  - Syringocele
- Low-lying conus

LEARNING OBJECTIVES

- Vertebral anomalies:
  - posterior spina bifida - wide or narrow
  - osteocartilaginous spur
Congenital defects of CNS that result from failure of the neural tube to close during first 30 days of fetal development
NEURAL TUBE DEFECTS: TYPES

- anencephaly
- encephalocele
- spinal dysraphism: defective closure of spinal neural tube
Spinal dysraphisms result from derangement in early embryonic development
- between 2 and 6 wk GA

Relevant steps
- Gastrulation (2–3 wk)
- Primary neurulation (3–4 wk)
- Secondary neurulation (5–6 wk)
Gastrulation: bilaminar disk is converted into a trilaminar disk
Primary neurulation

neural plate $\rightarrow$ neural fold $\rightarrow$ neural tube
Secondary neurulation

- forms tip of conus medullaris and filum terminale
- neural plate → solid cord → cavitation
CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

- skin covering

OPEN SPINAL DYSRAPHISM

CLOSED SPINAL DYSRAPHISM

subcutaneous mass

CSD WITH SUBCUTANEOUS MASS

CSD WITHOUT SUBCUTANEOUS MASS

SIMPLE

COMPLEX

Cutaneous birthmarks present in >50% of csds
- Angiomas, dimples, overgrowing hair, dyschromia, and dystrophy
CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

Open Spinal Dysraphism
- Myelomeningocele 99%
- Myelocele
- Hemimyelomeningocele
- Hemimyelocele

Closed Spinal Dysraphism
- with subcutaneous mass
- without subcutaneous mass

CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

Open Spinal Dysraphism

Closed Spinal Dysraphism

with subcutaneous mass
- Lipomas with dural defect
  Lipomyelomeningocele
  Lipomyelocele
- Myelocystocele
  Terminal
  Nonterminal
- Meningocele

without subcutaneous mass
CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

Open Spinal Dysraphism

Closed Spinal Dysraphism

with subcutaneous mass

without subcutaneous mass

SIMPLE
• Dermal sinus
• Intradural lipoma
• Filar lipoma
• Tight filum terminale
• Abnormally elongated spinal cord
• Persistent terminal ventricle

COMPLEX
• Dorsal enteric fistula
• Neurenteric cysts
• Diastematomyelia
• Caudal agenesis
• Segmental spinal dysgenesis
TERMINOLOGY: PLACODE

Definition: segment of non-neurulated embryonic neural tissue (i.e. frozen in neural plate stage)

Location along cord
- Terminal: at caudal end of cord
  - apical: defect involves apex of cord
  - parietal: defect involves longer segment of cord
- Segmental: at intermediate level along cord
Definition: clinical syndrome that may ensue

- As a complication of myelomeningocele repair
- As presentation of several forms of CSD
  - spinal lipomas
  - tight filum terminale
  - diastematomyelia
  - caudal agenesis
- In patients with filar lipomas & conus medullaris in "normal" position
TCS involves

- **progressive neurologic deterioration**
  - motor and sensory dysfunction of the lower extremities
  - muscle atrophy
  - decreased or hyperactive reflexes
  - urinary incontinence
  - spastic gait

- **orthopedic deformities**
  - scoliosis or foot and hip deformities
OPEN SPINAL DYSRAPHISM

- Myelomeningocele
- Myelocele
- Hemimyelomeningocele
- Hemimyelocele
Clinical dx:

- **Myelomeningocele:** elevation of neural placode by underlying expanded subarachnoid space
- **Myelocele:** neural placode flush with surface of back
Myelomeningocele with Chiari II (not shown)
Sagittal and axial T2 in 27 week fetus
OPEN SPINAL DYSRAPHISM: RARE

- Radiologic dx:
  - Hemimyelomeningocele
  - Hemimyelocele
- Closure defect affects one hemicord in diastematomyelia

CLOSED SPINAL DYSRAPHISM
WITH SUBCUTANEOUS MASS
Open Spinal Dysraphism

Closed Spinal Dysraphism

- with subcutaneous mass
  - Lipomas with dural defect
    - Lipomyelomeningocele
    - Lipomyelocele
  - Myelocystocele
    - Terminal
    - Nonterminal
  - Meningocele

- without subcutaneous mass
  - Lipomyelocele
LIPOMAS WITH DURAL DEFECT

Components

- lipoma-placode interface  
  - +/- intraspinal lipoma
- subcutaneous lipoma
- wide posterior spina bifida
- tethering of spinal cord
- +/- hydromyelia

Incidence: 16.4% of all CSDs
LIPOMAS WITH DURAL DEFECT

- Lipomyelocele vs. lipomyelimeningocele
  - Lipoma-placode interface: within or outside spinal canal
  - Size of subarachnoid space ventral to lipoma-placode interface: normal or enlarged
Case 1

At 1 day: right L5 hemivertebra
At 4.5 months. **Lipomyelocele**

- lipoma-placode interface is within spinal canal
- intraspinal lipoma (arrow)
- subcutaneous lipoma
- wide posterior spina bifida
- tethering of spinal cord
- hydromyelia
At 2 years. Parietal placode (arrows). Had surgery 4 months later to release tethered cord
At 2 years. L5 hemivertebra (red arrow), spina bifida (green arrow)
Lipomyelomeningocele

- placode: terminal apical
- placode-lipoma interface: off-midline & outside spinal canal (arrow)
Components

- "syringocele": cystic expansion of central canal
- "meningocele"
- wide spina bifida
- spinal cord tethering
- +/- subcutaneous lipoma
- +/- hydromyelia
- CSD with subQ mass, tethered cord
- Omphalocele
- Rt hydroureteronephrosis
At 4 months

- Syringocele
- Meningocele
- Hydromyelia
- Subcutaneous lipoma
At 4 months

- Syringocele (*red arrows*)
- Meningocele (*green arrows*)
- Hydromyelia (*blue arrows*)
- Subcutaneous lipoma (*yellow arrowhead*)
Hydromyelia (blue arrows) and syringocele (red arrows)
At day 1
What are the findings? Differential diagnoses?
Case 5

- “syringocele” and/or “meningocele”
- spina bifida
- spinal cord tethering
- hydromyelia
- + subQ lipoma

CSD with subcutaneous mass
- Lipomas with dural defect
  - Lipomyelomeningocele
  - Lipomyelocele
- Myelocystocele
- Meningocele

At day 1
What are the findings? Differential diagnoses?
At day 1. In neonate, findings may not be clear cut!
At 7.5 months. Syringocele and hydromyelia (arrows)
At 7.5 months. \textbf{(Lipo)myelocystocele} & partial diastematomyelia (arrows)
Case 6

At 1 day

What are the findings? Differential diagnoses?
Case 6

- “syringoele” and/or “meningocele”
- spina bifida
- spinal cord tethering
- hydromyelia
- subQ lipoma

At 1 day

What are the findings? Differential diagnoses?

- Lipomas with dural defect
  Lipomyelomenigocele
  Lipomyelocele
- Myelocystocele
- Meningocele
NONTERMINAL MYELOCYSTOCELE

Components
- meningocele with a thin neck
- narrow posterior spina bifida
- contents of sac
  - expanded hydromyelic cavity lined by posterior wall of spinal cord
- OR
- thin fibroneural stalk that attaches to dome of meningocele

Nonterminal Myelocystocele
Nonterminal Myelocystocele
Thin stalk (*arrows*) corresponded to a fibroneural filament at surgery
Nonterminal Myelocystocele
Thin stalk (*arrows*) corresponded to a fibroneural filament at surgery
Case 8 - fetus

Thoracic limited dorsal myeloschisis
21 wk, 5 d fetus
Case 8 - neonate

Sagittal and axial T2 SPACE
MENINGOCELE

Definition: herniation of a CSF-filled sac, lined by dura and arachnoid, through a posterior spina bifida

- may contain nerve roots and, more rarely, a hypertrophic filum terminale
- no part of the spinal cord is contained within sac
MENINGOCELE

- Incidence: 2.4% of all CSDs
- Types of meningoceles
  - posterior: CSD with subcutaneous mass
  - anterior: CSD without subcutaneous mass
  - intrasacral: CSD without subcutaneous mass
Case 9
Intrasacral meningocele

T2

T2

CLOSED SPINAL DYSRAPHISM
WITHOUT SUBCUTANEOUS MASS
SIMPLE
CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

Open Spinal Dysraphism

Closed Spinal Dysraphism

SIMPLE
• Dermal sinus
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COMPLEX
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• Caudal agenesis
• Segmental spinal dysgenesis

with subq mass

without subcutaneous mass
FILAR LIPOMA AND FATTY FILUM

- Adipose infiltration of whole length or part of filum terminale
- Incidence: 3.7%
Case 10

Fatty filum
Case 11

Filar lipoma
Premature disjunction between neuroectoderm and cutaneous ectoderm →

- Mesenchyme gains access to interior of neural tube →
- Induced to differentiate into fatty tissue
SPINAL LIPOMA

- Subpial, contained within an intact dural sac
- Lies in groove formed by unapposed folds of placode
- Large lipomas may displace cord laterally
  - off-midline placode-lipoma interface
- Rare: completely intramedullary lipomas or diffuse medullary lipomatosis
Case 12

Intradural lipoma *(arrows)*, and tethered cord
Case 13

At 2 days. Intradural lipoma (arrows)
At 7 weeks. Intraspinal mass has grown
Comment on lipoma growth with child growth
At 7 weeks. Large intradural lipoma (arrows) displaces cord laterally.
Post partial resection, complicated by abscess
Conus seen at lumbar level (arrow)
Cases 14 & 15

Lumbar intradural lipoma tethering cord

Intradural lipoma and hemangioma tethering cord
Case 16

Thoracic intradural lipoma
DORSAL DERMAL SINUS

- Definition: epithelium-lined fistula that extends inward from skin surface to a variable depth
- +/- pierce dura to reach intradural compartment and end in
  - subarachnoid space
  - hypertrophic or fibrolipomatous filum terminale
  - low-lying conus medullaris
  - intraspinal lipoma
- +/- dermoids (11%)
DORSAL DERMAL SINUS: CLINICAL EXAM

- Midline dimple or pinpoint ostium
- +/- hairy nevus, capillary hemangioma, dyschromic area
- Location of opening
  - Above intergluteal cleft
  - Usually directed inferiorly
  - Lumbosacral, cervical, thoracic, occipital
- Complications: local infection, meningitis, abscess
Case 17

14 month old female. **Dermal sinus and infected dermoid cyst** (arrows); hydromyelia
2 year old female. Dermal sinus and infected dermoid cyst (arrows)
2 year old female. **Dermal sinus** (arrow) and **dermoid cyst**, infected.
CLOSED SPINAL DYSRAPHISM
WITHOUT SUBCUTANEOUS MASS

COMPLEX
• Dorsal enteric fistula
• Neurenteric cysts
• Diastematomyelia
• Caudal agenesis
• Segmental spinal dysgenesis
COMPLEX DYSRAPHIC STATES

Disorders of midline notochordal integration

- Result in longitudinal splitting

  - Types
    - Dorsal enteric fistula/sinus
    - Neurenteric cyst
    - Diastematomyelia

Disorders of notochordal formation

- Result in absence of a notochordal segment

  - Types
    - Caudal agenesis
    - Segmental spinal dysgenesis
NEURENTERIC CYSTS

- Intraspinal counterpart of gut duplications
- Intraspinal, typically intradural
- Location: cervicothoracic, lumbar, posterior fossa
  - anterior to cord, posterior to cord, intramedullary
- Solitary/associated with anterior/posterior spinal dysraphism (including diastematomyelia)
Case 19

Neurenteric cyst, intradural extramedullary with block vertebrae

13 year old male with left leg weakness and hyper-reflexia

Sagittal and axial T2
Neurenteric cyst, intradural extramedullary with block vertebrae
Definition: separation of spinal cord into 2 halves

- Usually symmetrical splitting
- Complete/partial splitting
- Within one/two dural tubes
- Hemicords separated by
  - osteocartilaginous spur; complete/incomplete spur
  - fibrous septum
  - nothing
  - +/- hydromyelia
  - Usually hemicords fuse below spur
DIASTEMATOMYELIA: TYPES & INCIDENCE

Split cord malformation

- Pang type 1: separate dural sacs with spur (25%)
- Pang type 2: single dural sac without spur (75%)

Incidence: 3.8% of CSDs
Type I, osteocartilaginous spur

Type II, with fibrous septum

Type II, without septum

Type II, partial splitting
Type I, complete osseous spur

Type II, with fibrous septum

Type II, without septum

Type II, partial splitting

Type I, incomplete osseous spur

Case 20

Type II, partial splitting
Case 21

Type I, complete osseous spur (arrows)
Type I, complete osseous spur (arrows)
Hemicords do not fuse distal to spur and left hemicord ends in an intradural lipoma (arrows)
CAUDAL AGENESIS

- **Definition**: heterogeneous constellation of anomalies comprising
  - total or partial agenesis of caudal spinal column
  - anal imperforation
  - genital anomalies
  - bilateral renal dysplasia or aplasia
    - pulmonary hypoplasia
  - lower limbs usually are dysplastic
    - fusion or agenesis results in most severe cases (sirenomelia)

- **Incidence**: 16.3% of all CSDs
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AGENESIS OF THE SACROCOCCYGEAL SPINE IN SYNDROMIC COMPLEXES

- **OEIS:** Omphalocele, cloacal Exstrophy, Imperforate anus, and Spinal deformities
- **VACTERL:** Vertebral abnormality, Anal imperforation, Cardiac anomalies, TracheoEsophageal fistula, Renal abnormalities, Limb deformities
- **Currarino triad:** partial sacral agenesis, anorectal malformation, and presacral mass (teratoma and/or meningocele)
AGENESIS OF THE SACROCOCCYGEAL SPINE, ASSOCIATIONS

- Lipomyelomeningocele and terminal myelocystocele: in 20%
- Maternal diabetes mellitus
  - 1% of offspring of diabetic mothers
Case 22

CA type I & lipomyelocele. 4 day old
Last vertebra is a dysplastic L3. Ilioiliac approximation (arrows).
• intraspinal lipoma (red arrow)
• subcutaneous lipoma (green arrow)
• posterior spina bifida
• lipoma-placode interface is within spinal canal (blue arrow)
CAUDAL AGENESIS: TYPES

Based on location and shape of conus medullaris

- Type I: high and abrupt
- Transitional type I/II
- Type II: low and tethered
CAUDAL AGENESIS: TYPES I, I/II

- **Type I: high and abrupt**
  - last vertebra: L5-S2
  - spinal terminus: wedge-shaped, @ T12-L1
  - “double-bundle shape”: separation of the anterior and posterior spinal roots of cauda equina

- **Transitional type I/II**
  - last vertebra: S5 (coccygeal agenesis)
  - spinal terminus: wedge-shaped, @ normal level
Case 23

CA Type I, wedge-shaped spinal terminus at L1
CA Type I, “double-bundle shape” cauda equina nerve roots (arrows)
CAUDAL AGENESIS: TYPE II

Type II: low and tethered

- last vertebra: up to S4 may be present
- spinal terminus
  - partial agenesis of caudally stretched conus
  - tethered to
    - tight filum
    - lipoma
    - terminal myelocystocele
    - lipomyelomeningocele
    - caudal tumor (germ cell tumor)
    - neck of an anterior sacral meningocele
CA Type II, tight filum
SAM Questions
Sagittal T2-W and T1-W images of a 4-month old with a spinal dysraphism are shown. This anomaly may be classified as a:

a) open spinal dysraphism  
b) closed spinal dysraphism with subcutaneous mass  
c) closed spinal dysraphism without a subcutaneous mass, simple type  
d) closed spinal dysraphism without a subcutaneous mass, complex type
Question 2

Sagittal T2-W and T1-W images of a 4-month old with a spinal dysraphism are shown. The arrows point to a

a) meningocele
b) cystocele
c) hydromyelia
d) syringocele
Sagittal T2-W and T1-W images of a 4-month old with a spinal dysraphism are shown. The diagnosis is

a) lipomyelomeningocele  
b) terminal lipomyelocystocele  
c) myelomeningocele  
d) lipomyelocele
CLINICAL-NEURORADIOLOGICAL CLASSIFICATION

SPINAL DYSRAPHISM

- skin covering

OPEN SPINAL DYSRAPHISM

CLOSED SPINAL DYSRAPHISM

subcutaneous mass

CSD WITH SUBCUTANEOUS MASS

CSD WITHOUT SUBCUTANEOUS MASS

SIMPLE

COMPLEX

SUMMARY

- Spinal cord development in fetus
- Spinal dysraphisms, classification
  - Open spinal dysraphisms
  - Closed spinal dysraphisms
    - with subcutaneous mass
    - without subcutaneous mass
    - simple
    - complex
THANK YOU!
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