Imaging Perinatal Spinal Dysraphisms: Clinical Conundrums and Diagnostic Dilemmas

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Disclosure

- McDonald: nothing to disclose
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Imaging Perinatal Spinal Dysraphisms

Educational Goals

• To review the appearance of the fetal spine during development using both ultrasound and MRI, with attention to normal anatomic variants

• To discuss the imaging hallmarks of the most common spinal malformations encountered in the fetal setting, including:
  • Myelocoele and myelomeningocele
  • Lipomyelomeningocele/intraspinal lipoma
  • Caudal regression and sacral agenesis
  • Segmental dysgenesis and other bony malformations
  • Syndromic constellations associated with dysraphism

• To correlate prenatal findings with postnatal MRI and clinical outcomes
Imaging Perinatal Spinal Dysraphisms: An Evolving Field

- Spinal dysraphisms (SD) or neural tube defects (NTD) affect 1 out of 1000 live births in the US\textsuperscript{1}
  - In-utero MMC repair is currently one of the few CNS lesions amenable to fetal intervention.

Prenatal surgery for myelomeningocele reduced the need for shunting and improved motor outcomes at 30 months, but was associated with both maternal and fetal morbidity \textsuperscript{2}

- Advanced techniques including fetal MR and 3-D US may be required routinely to accurately classify these spinal dysraphisms.
Imaging Perinatal Spinal Dysraphisms: US Technique

- **Screening**
  - Biochemical testing (maternal AFP) – elevated in open NTD but not skin covered closed NTD
  - Prenatal 2-D US
    - Accuracy variable based on experience, fetal lie and maternal habitus

- **Targeted US**
  - Accuracy up to 100% in the setting of elevated maternal AFP
  - 3-plane evaluation and correlation with intracranial findings for associated anomalies (skull shape, ventricular size, etc)
  - High frequency linear transducers to delineate cord position/tethering, placode contents, presence of scoliosis or associated vertebral anomalies
    - Can determine level of a dysraphism within one vertebral body
Imaging Perinatal Spinal Dysraphisms: MRI Technique

• Useful adjunct for further delineation of anatomy, musculature or brain anomalies

• Technique
  • 1.5 T using a torso, body or cardiac phased array surface coil
  • Large FOV to document lie, placentation and amniotic fluid volume
  • Subsequent smaller FOV in orthogonal planes with respect to fetal spine and brain
    • T2 weighted SSFSE or HASTE at minimal thickness (2-4 mm) are most useful in the assessment of spine and brain anomalies
    • Adjunct fast T1 to assess fat and blood content
    • EPI for skeletal and vascular structures as needed
  • Axial plane of particular importance for evaluating cord tethering and placode location
  • Sagittal and coronal abdominal planes for the evaluation of scoliosis and associated GI/GU anomalies
Representative second trimester coronal (left) and sagittal (right) large FOV SSFSE images during fetal MRI. Note cephalic fetal lie (arrow, right) and posterior, fundal placenta (arrowheads).
Normal fetal spine – ultrasound and MRI

T2 weighted small FOV SSFSE sagittal image of the fetal spine during the second trimester

High frequency, linear probe sagittal view of the thoracolumbar spine during the second trimester
Normal fetal spine – ultrasound and MRI

T2 weighted SSFSE sagittal image of the fetal spine at 22 weeks

High frequency US cervical spine

Lateral process ossification center

trachea
Normal fetal spine – ultrasound and MRI

T2 weighted SSFSE sagittal image of the fetal spine at 22 weeks

High frequency US thoracic spine

Vertebral ossification center

rib
Normal fetal spine – ultrasound and MRI

T2 weighted SSFSE sagittal image of the fetal spine at 22 weeks

High frequency US lumbar spine

Vertebral ossification center
Several studies have examined the role of 3D ultrasound in evaluation of the fetal spine as a problem solving tool.

- Main advantage: possibility of visualizing the entire length of the bony elements of the spine on a single image.
  - Most useful in the assessment of sacral agenesis, segmental dysplasias.

- Pitfall: neural tube defects.
  - Ossification centers may appear grossly normal with only subtle splaying of the lateral elements as evidence of underlying neural tube defect.

Sagittal “start” scan

C-plane movement (posterior to anterior)

Fetal spine “up”

Lateral processes

Maximum mode: all three ossification centers

Adapted from Gilu et al 2011.

Imaging Perinatal Spinal Dysraphisms: 3-D Ultrasound
Suspected thoracic segmental anomaly at 14 weeks on 2D high frequency linear imaging.

Additional evaluation with 3D ultrasound demonstrated normal cervical, thoracic and lumbar spine anatomy without evidence of segmentation anomalies.
Known lumbosacral open neural tube defect in second trimester fetus (above, arrow).

Concomittant imaging with 3-D ultrasound does not clearly identify open neural tube defect.
Four principal processes occurring in sequential order:

- Gastrulation with development of the notochord
- Primary neurulation
- Segmentation with appearance of the somites
- Secondary neurulation
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Adapted from Ruefener et al 2010
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Four principal processes occurring in sequential order:

- Gastrulation with development of the notochord
- Primary neurulation
- Segmentation with appearance of the somites
- Secondary neurulation – sacral spine

Adapted from Ruefener et al 2010.
Tortori-Donati classification

Open dysraphism (no skin covering)
- Myelocele
- Myelomeningocele
- Hemimyelocele
- Hemimyelomeningocele

Closed dysraphism (skin covering)
- Associated subcutaneous mass?
  - yes
    - Lipomyelocele/-meningocele
    - Myelocystocele
    - Meningocele
  - no
    - Tight filum terminale
    - Intradural lipoma
    - Dermal sinus
    - Neurenteric cyst
    - Caudal regression
    - Segmental dysgenesis
Case #1

34 F G2P1 presenting for second trimester anatomy scan.

Beaking of the frontal bones (“lemon” shaped skull, arrows near right) with crowding of the posterior fossa (far right, arrowheads) suggests underlying Chiari II malformation. Also noted was bilateral ventriculomegaly (9 mm, near right) with choroidal separation.

Sagittal (left) and transverse (right) ultrasound demonstrating a large posterior dysraphism extending from the lower thoracic spine to the sacrum (arrows) containing elements of the neural placode and nerve rootlets (arrowhead).
34 F G2P1 presenting for second trimester anatomy scan with suspected Chiari II and myelomeningocele.

Sagittal (left) and axial (top) SSPSE fetal MRI redemonstrating an open neural tube defect containing CSF and neural elements extending from L2 to S2 (left, arrows). There is associated splaying of the lamina at these levels with absent posterior elements (top, arrowheads). Crescenteric region of T2 hypodensity along the dorsal margin of the defect likely represents part of the neural placode (asterisk).

Case #1

Chiari II. Sagittal SSFPSE also reveals a small posterior fossa with cerebellar tonsillar herniation of ~3 cm below the foramen magnum (arrowhead, top). Biconcavity of the fetal calvarium is again seen on axial SSFPE (near right, arrowheads) with complete lack of the normal subarachnoid spaces and bilateral ventriculomegal (axial GRE, far right, arrows).
Open neural tube defect: Myelomeningocele (MMC)

- **Etiology:** Primary neurulation
  - Defective closure of the neural tube results in a placode that fails to detach from the adjacent surface ectoderm

- **Imaging:**
  - Lumbosacral > thoracic > lumbar > cervical
  - US: flared laminae, loss of posterior epidural fat at anomaly level, low lying cord/roots
  - MRI: flattened midline neural placode pushed dorsal +/- adjacent bone and muscle malformations

- **Associated anomalies**
  - Chiari II - incomplete/defective expansion of the rhombencephalic vesicle secondary to chronic CSF leakage
  - Also diastatomyelia, dermal sinus

- **Prognosis**
  - Stable neurological defect as best outcome
  - Hydrocephalus and tethered cord determine future deterioration

- **Pearls:** always image for concomitant CNS abnormalities

Adapted from Ruefener *et al* 2010.
Case #2

27 F G1P0 presenting for second trimester anatomy scan.

Axial views of the sacral spine (far left) demonstrating an open neural tube defect (arrow). Note splaying of the bilateral lamina (arrowheads). The more proximal lumbar spine is normal in appearance.

Crowding of the posterior fossa (right) with flattening of the cerebellar contour (arrows), the so-called “banana sign” suggesting coexisting Chiari II malformation.
27 F G1P0 with second trimester anatomy scan concerning for open neural tube defect and associated Chiari II malformation.

Sagittal (top left) and axial (bottom left) SSFSE views demonstrating an open neural tube defect extending from L5-S2 (arrows). On axial view, the neural placode appears flush with the skin surface (arrowheads) without obvious associated protruding meningeal sac.

Sagittal (top) SSFSE demonstrating coexisting Chiari II malformation with inferior herniation of the cerebellar tonsils into the foramen magnum (arrow).
Open neural tube defect: Myelocele (MC)

- **Etiology:** Primary neurulation
  - Defective closure of the neural tube results in a placode that fails to detach from the adjacent surface ectoderm
  - <2% of all OSDs

- **Imaging:**
  - Key anatomic feature is flattened midline neural placode flush with the adjacent skin, rather than pushed dorsally as in MMC

- **Associated anomalies:**
  - Chiari II, dermal sinus, diastatomyelia, vertebral dysgenesis/anomalies

- **Prognosis:**
  - Similar to MMC

Adapted from Rufener et al 2010.
Classification

Open dysraphism (no skin covering)
- Myelocele
- Myelomeningocele
- Hemimyelocele
- Hemimyelomeningocele

Closed dysraphism (skin covering)
- Associated subcutaneous mass?
  - yes
  - Lipomyelocele/-meningocele
    - Myelocystocele
    - Meningocele
Case #3

16 F with lower extremity weakness and neurogenic bladder.

Sagittal (left) and axial (right) T2 weighted images demonstrating closed neural tube defect extending from L3 to L5 (arrowheads). A tethered, low lying conus is demonstrated at the level of L3/4 (arrows) adjacent to a fat density within the central canal (asterisk). A large posterior lipoma is present (open arrows, left).
Closed spinal dysraphism with subcutaneous mass: lipomas with a dural defect (lipomyelocele/lipomyelomeningocele)

- **Etiology:** defect in primary neurulation
  - Mesenchymal cells migrate through a dural defect into the neural tube and form lipomatous tissue

- **Imaging:**
  - US: can be subtle secondary to absent intracranial findings and intact skin
    - Best clue: echogenic mass
  - MRI: subcutaneous fatty mass above the intergluteal crease + placode lipoma interface
    - Inside the spinal canal = lipomyelocele
    - Outside the spinal canal = Lipomyelomeningocele

- **Pearls:** always look for associated dysraphism when an intraspinal mass is present. US can also detect associated cord tethering, hydromyelia and syringomyelia.

Adapted from Ruefener et al 2010.
Case #4

33 G4P1 at 19 weeks referred for lumbar mass on screening ultrasound

Left. Axial (top) and coronal (bottom) views of the lumbosacral spine demonstrating a large fluid filled sac extending from a defect in the posterior elements at L4/5 (arrows). Thin internal septations are seen within the dorsal sac (arrowhead).

Top right. Normal intracranial findings without evidence of crowding of the posterior fossa, hydrocephalus or abnormal frontal bone contour.
Case #4

33 G4P1 at 19 weeks referred for lumbar mass on screening ultrasound

Sagittal (far left) and axial (left) SSFSE images demonstrate a 1 cm fluid signal intensity collection in the posterior lumbosacral soft tissues (arrows). A thin linear septation is appreciated internally (arrowhead), presumed nerve root.

Axial (far right) and sagittal (right) SSFSE imagines confirming normal neuroanatomy without evidence of cerebellar herniation or ventricular dilation to suggest associated Chiari II malformation.
Closed spinal dysraphism with subcutaneous mass: meningocele

- **Etiology:** unknown
- **Imaging:**
  - Most commonly lumbosacral but can be anywhere along the dorsal canal
    - Anterior less common, usually presacral
  - Key determination: presence of neural elements
  - **US:** CSF-filled sac protrudes through posterior defect +/- associated conus abnormalities
    - Primary utility for screening fetal US, less useful in older children due to spinal ossification
  - **MRI:** skin-covered dural sac without or without elements of the filum terminale/nerve roots
- **Associated Chiari II rare**
- **Prognosis:**
  - Better than MMC and MC
    - Asymptomatic patients warrant conservative observation
    - Surgical resection and dural repair if symptomatic

Adapted from Veenboer et al 2015
Classification

Open dysraphism (no skin covering)

Myelocoele
Myelomeningocele
Hemimyelocoele
Hemimyelomeningocele

Closed dysraphism (skin covering)

Associated subcutaneous mass?

- no

Tight filum terminale
Intradural lipoma
Dermal sinus
Neurenteric cyst
Caudal regression
Segmental dysgenesis
Case # 5

Limited access to prenatal care. 1 month old male with lower extremity hypotonia.

Sagittal T1 (far left) demonstrates a low lying conus terminating at the level of L3 (arrow). Axial T1 (bottom right) and T2 (top right) FSE sequences reveal thickening of the filum with internal T1/T2 hyperintensity, consistent with fat density (arrowhead).
Closed spinal dysraphisms without subcutaneous mass: Tethered cord/tight filum terminale

- **Imaging:**
  - **US:** best tool for screening
    - Low lying conus (lower than the inferior margin of T2) +/- thickening filum
    - Reduced or absent spinal cord movement
  - **MRI:** used to confirm positive ultrasound
    - T1 – thickened filum (>2mm) +/- hyperintense lipoma
    - T2 – secondary syringohydroyelia or myelomalacia seen in up to 25%. +/- chemical shift artifact from fatty holum

- **Associations:** cutaneous stigmata, VACTERL, open or closed spinal dysraphism, incomplete fusion of the posterior elements (nearly all)

- **Prognosis:**
  - Progressive irreversible neurological impairment if untreated
  - Majority of patients show improvement or stabilization of deficits with surgical untethering

Longitudinal ultrasound (above) demonstrating a low lying conus terminating at L5 (arrow). On MRI (left) the filum appears thickened (arrow).
30 F presenting for lumbar spine MRI secondary to chronic low back pain.

Focal, ovoid intradural, extramedullary lesion adjacent to the conus at the L1/L2 interspace (arrows) without overlying dysraphism of the posterior elements (arrow head, bottom right). Note chemical shift artifact on T2 phase images (center, top) confirming internal macroscopic fat density.
Closed spinal dysraphisms without subcutaneous mass: intradural lipoma

- **Etiology:** primary neurulation defect
  - Surround mesenchymal precursors enter the central spinal canal, impeding closure of the neural folds

- **Imaging**
  - **US:** Hyperechoic intraspinal mass
  - **MRI:** intradural, extramedullary lesion following fat on all sequences +/- localized spinal dysraphism

- **Prognosis:**
  - Conservative if asymptomatic
  - Can grow rapidly during infancy; surgical resection and/or untethering of the cord with good long term outcomes

- **Pearls:** use of chemical fat saturation or inversion recovery MRI can confirm fat content
Case # 7

1 month old male ex 35 week premature delivery with multiple congenital abnormalities.

Sagittal (left), coronal (top) and axial (top right) T2 weighted images demonstrating a small, multilobulated cystic structure (arrows) extending from the central canal into the posterior mediastinum (arrows). The cystic structure is isointense to CSF. Note associated segmental anomaly/scoliosis of a midthoracic vertebral body (arrowheads).
Closed spinal dysraphisms without subcutaneous mass: neurenteric cyst

- **Etiology:** subgroup of split notochord syndrome spectrum
  - Sporadic or syndromic (Klippel-Feil, VACTERL, OEIS syndromes)

- **Imaging**
  - **US:** Hypoechoic intraspinal/paraspinal cyst
  - **MRI:** Well-circumscribed, fluid intensity cystic lesion
    - Hypo- → isointense (to CSF) depending on protein/mucin content
    - ± cord myeloschisis, focal cord atrophy
  - **Nuc med:** Tc-99m cyst uptake (gastric mucosa) confirms diagnosis

- **Prognosis:**
  - Variable (asymptomatic to progressive neurological deterioration)
  - Most patients show clinical improvement after resection

Sagittal (top) and axial T2 images demonstrating bilobed neurenteric cyst extending from the epidural space into the posterior mediastinum.
Case # 8

27 F G1P0 presenting for evaluation of abnormal spine on second trimester screening ultrasound from outside institution.

Top left. High frequency longitudinal ultrasound demonstrating segmental anomalies were seen at the level of T10 and T12/L1. On 3D images (left) the rib appears fused at 9-10 and 11-12 (arrow heads). Marked leftward scoliosis is seen at the T12-L1 level.

Left. The conus medullaris (distal part of the spinal cord) appears to be at the L3-4 level rather than the L2 level suggesting a tethered cord.
Closed spinal dysraphisms without subcutaneous mass: segmental dysplasia

- **Etiology:**
  - Complex possibly representing inappropriate apoptosis during gastrulation vs abnormal primary neurulation

- **Imaging**
  - US: most useful in prenatal setting
  - MR: vertebral anomalies +/- low lying conus/tethered cord, absent nerve roots at level of the lesion

- **Pearls:** check for associated anomalies including open/closed NTD, visceral (renal, cardiac, bladder) and orthopedic (clubfoot, etc)
Case #9

42 F G5P3 with poor controlled diabetes type II (HbA1c 11.6%) presenting for second trimester anatomy scan with concern for “short spine” on ultrasound from outside hospital.

Sagittal and axial prenatal ultrasound demonstrating poor ossification of the thoracolumbar spine (top) with truncation of the distal lumbar/sacral elements (top, arrow). Hypoplastic vs. absent iliac bones and sacrum (left, white arrows) suggest caudal regression syndrome.
Closed spinal dysraphisms without subcutaneous mass: caudal regression syndrome

- **Etiology:** complex
  - Varying degrees of developmental failure involving the sacral and lumbar spine caused by insult prior to 4th gestational week

- **Imaging**
  - **US**
    - first trimester short CRL
    - Second trimester: abrupt spine termination on longitudinal views
    - “shield” appearance of the iliac wings
  - **MR**
    - Best modality for evaluating associated anomalies of the spinal cord and conus

- **Pearls:** always check for spine ossification centers in axial scan at the level of iliac wings
Prenatal imaging with US and MRI allows for precise examination of the spinal canal and aids the radiologist in the identification of significant pathologic conditions.

Detection is predicated upon a complete understanding of the normal developmental anatomy of the spinal cord and associated structures, as well as a familiarity of the imaging hallmarks of dysraphism on both US and MRI.

Early and accurate diagnosis facilitates appropriate clinical management, ranging from birth planning to neonatal surgical techniques, which reduces patient complications and improves overall clinical outcomes.
References

Thank you for visiting exhibit # eEdE-217

For questions or comments, please contact:
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