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REVIEW ARTICLE **CME**

Diagnostic Flowchart and Imaging Features of Common Congenital Anomalies of the Neonatal Spine

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ABSTRACT

Paediatric spinal pathologies are commonly encountered in clinical practice. This article summarises the normal appearance of lumbar ultrasonography (LUS) and the imaging techniques used to examine the spine in neonates. The last section of this article is a pictorial illustration of ultrasonography and magnetic resonance imaging (MRI) features of common paediatric spinal pathologies. These include filar cyst, arachnoid cyst, filar fibrolipoma, low-lying cord, syringomyelia, myelomeningocele, Chiari malformations and sacrococcygeal teratoma. Current literature and guidelines on the indications for imaging and available modalities for investigation of occult spinal dysraphism are also reviewed. Isolated simple sacral dimple is no longer an indication for neonatal LUS. Indications for LUS include other cutaneous stigmata, abnormal antenatal ultrasound, and low-risk congenital anomalies. For neonates presenting with neurological deficit, discharging lesion or high-risk congenital anomalies, MRI is indicated. This review also proposes a diagnostic flowchart for better triage of patients to the appropriate modality of imaging in order to achieve satisfactory diagnostic accuracy and enhance cost-effectiveness.

Key Words: *Magnetic resonance imaging; Pediatrics; Spinal dysraphism; Spine; Ultrasonography*

中文摘要

新生兒常見的先天性脊柱畸形：診斷流程和影像學特徵

王先民、王嘉文、蘇妙兒、梁懿芳、廖健雄、朱昭穎

小兒脊柱病變於臨床實踐中經常遇見。本文總結了腰椎超聲檢查（LUS）的正常表現和用於檢查新生兒脊柱的影像技術。文末章節圖解說明常見小兒脊柱病症的超聲和磁共振成像（MRI）特徵。這些病症包括：絲狀囊腫、蛛網膜囊腫、絲狀纖維脂瘤、脊髓低位、脊髓空洞症、脊髓脊膜膨出、Chiari氏畸形及骶尾部畸胎瘤。本文亦對當前隱性椎管閉合不全影像檢查指徵和現有診斷方式指徵的相關文獻和指南作一回顧。孤立單發的骶骨凹不再是新生兒LUS指徵。LUS指徵包括皮膚紅斑、產前超聲結果異常和低度危險的先天性畸形。如新生兒出現神經功能缺損、排泄障礙或高度危險的先天性畸形，便有需要進行MRI。為達至令人滿意的診斷準確率及提高成本效益，本文亦提出了一診斷流程圖以便能更好地把病人分流出去接受適宜的影像檢查。

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INTRODUCTION

Lumbar ultrasonography (LUS) of the neonatal spine is commonly performed to assess potential occult spinal dysraphism (OSD) and lumbosacral soft tissue masses.[1] The term dysraphism refers to incomplete closure of a raphe, defective fusion, particularly of the neural tube. In this review, we present the technique of LUS to examine the neonatal spine, its normal sonographic appearance, and imaging features encountered in common paediatric spinal pathologies, especially OSD. We also review the recent literature on the indications for imaging and recommend appropriate modalities for the detection of OSD.

TECHNIQUE OF LUMBAR ULTRASONOGRAPHY IN NEONATE

In our institution, we perform LUS of the spinal cord in neonates with an IU22 scanner (Philips Healthcare, Best, The Netherlands) equipped with a high resolution 5- to 12-MHz linear-array transducer. Preferably, the infants are scanned in the prone position with their bodies curved over a pillow and head mildly elevated. This allows cerebrospinal fluid (CSF) to accumulate low down in the spinal canal, hence optimising visualisation. It also enables accentuation of lumbar lordosis and better defining of the lumbosacral junction. This position also produces a better acoustic window with splaying of the spinous processes. Alternatively, a decubitus position can be adopted in order to calm a struggling baby with bottle or breast feeding. Images are obtained in the longitudinal and transverse planes. Firstly the vertebral level is determined by counting up from the L5-S1 junction or the tip of coccyx. Then the level of the conus medullaris is determined. Thereafter, we aim to visualise the normal motion of the cord and nerve roots. If an intraspinal mass is evident, it can also be evaluated. The thickness of the filum terminale is also measured (normal, <2 mm). The integrity of the posterior elements of the lumbar spine is also assessed. Finally, if the clinical indication is sacral dimple, the

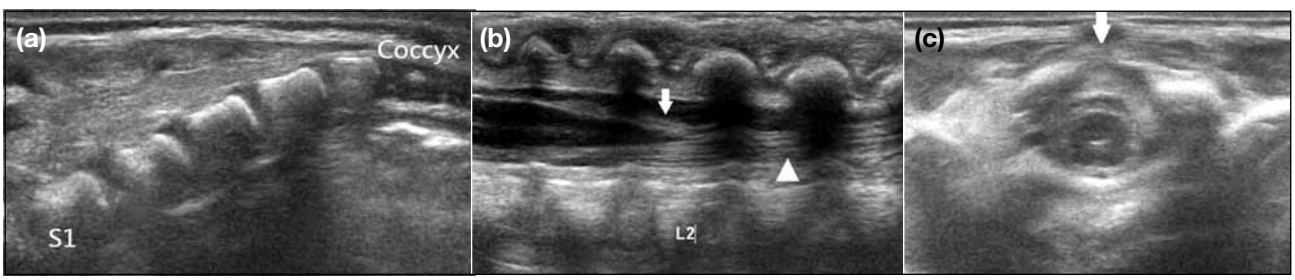

Figure 1. Normal neonatal lumbar ultrasonography: (a) a longitudinal image of the sacral region shows the normal S1-5 and coccygeal vertebrae. (b) A longitudinal image of the lumbar region shows the normal conus medullaris at the level of L2 (arrow) and the cauda equina (arrowhead). On real timing imaging, motion of the cord and nerve roots with respiration should be seen. (c) A transverse image of the lumbar region shows the normal appearance of the conus medullaris and the intact posterior vertebral element (arrow).

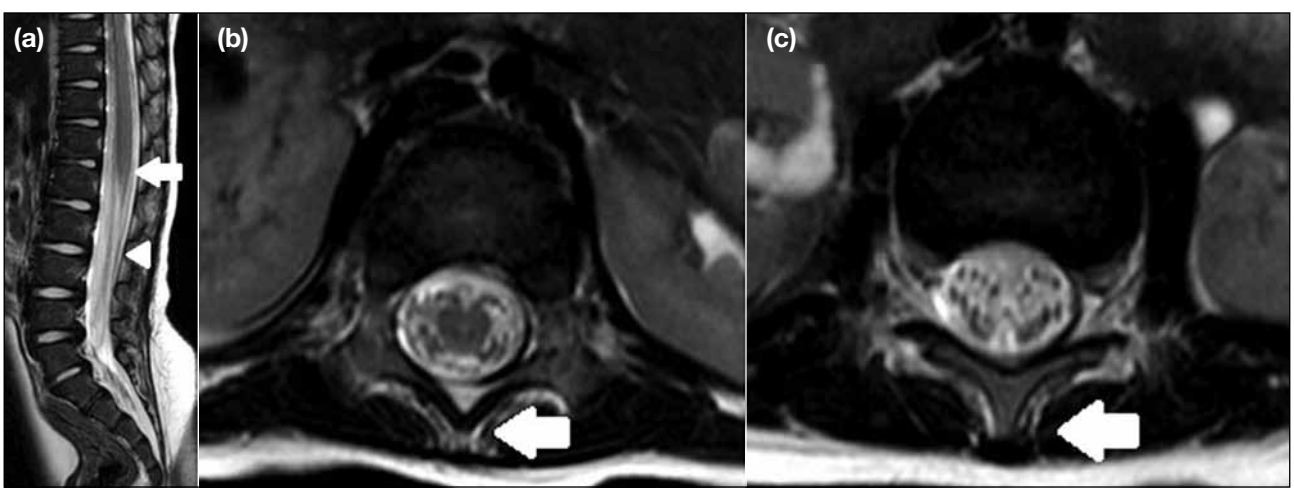

Figure 2. Normal T2-weighted magnetic resonance imaging (MRI) of the lumbosacral spine: (a) A sagittal image shows the normal appearance of the conus medullaris (arrow) at level of L1 and cauda equina (arrowhead). Axial images show (b) the conus medullaris and (c) the cauda equina at a lower level. Note the intact posterior vertebral elements (arrows).

corresponding area is closely examined to look for the presence of any subdermal tract extending to the spinal canal.

LUMBAR ULTRASONOGRAPHY OF THE NORMAL NEONATAL SPINE

The following is the checklist for a normal LUS examination of the neonate[2,3]:

1. Normal position of the conus medullaris: the level of termination of cord ranges from T12 to L3 (Figures 1 and 2); 98% of neonates have the conus medullaris at L2-L3 or higher. If it is at / below the L3 level, it is considered low lying.[4]
2. Motion of the cord and nerve roots with respiration: the cord and nerve roots show minimal motion with respiration. If no motion is evident, underlying cord tethering should be suspected.
3. Filum terminale: normally it is <2 mm in thickness, and may occasionally appear more echogenic.
4. Absence of any intraspinal mass: any hyperechoic intraspinal mass should be excluded to rule out the possibility of an intraspinal lipoma.
5. Integrity of the posterior element: subtle posterior defects may represent underlying spina bifida occulta. However, lack of ossification of the posterior element of the neonatal spine can sometimes make assessment difficult.
6. Absence of any subdermal tract in the sacral region.

NORMAL VARIANT ADEQUATELY INVESTIGATED BY ULTRASOUND ALONE

Filar Cyst

This is a well-defined fusiform midline cystic lesion within the filum terminale, just below the conus (Figure 3).[5,6] It is a normal variant, which on its own has no known clinical significance and does not warrant further imaging. Its midline location within the filum terminale is important to establish, and to differentiate it from other cystic intraspinal mass lesion.

COMMON PAEDIATRIC SPINAL ANOMALIES INVESTIGATED BY ULTRASOUND AND / OR MAGNETIC RESONANCE IMAGING

Arachnoid Cyst

Similar to its counterpart in the brain, this is a benign well-defined CSF-filled extra-axial cystic lesion in the spinal canal. It is commonly found in the dorsal aspect of the thoracic cord, due to a dural defect / adhesion that can be congenital or secondary to previous trauma or infection.[7] It is usually incidental, but if large, it may have a mass effect on the cord. Ultrasound typically demonstrates a well-defined fusiform extramedullary cystic lesion at the dorsal aspect of the thoracic cord

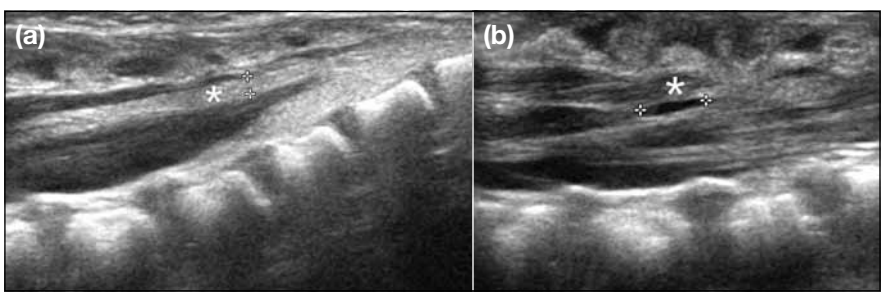

Figure 3. Longitudinal sonographic images of a neonatal spine show (a) prominent filum terminale (asterisk) measuring 1.6 mm thick, and (b) a small (5 mm) well-defined fusiform-shaped cystic lesion (asterisk) in the filum terminale, compatible with a small filar cyst. Note its midline location is just below the conus medullaris, which distinguishes it from other cystic intraspinal lesions.

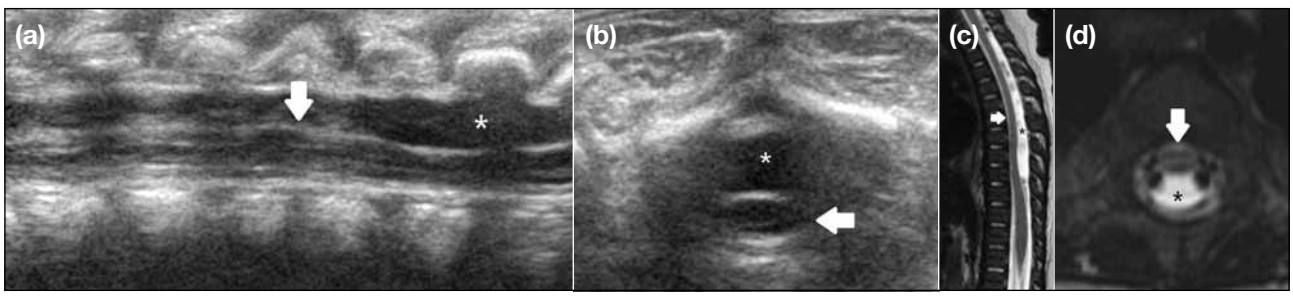

Figure 4. (a) Longitudinal, and (b) transverse ultrasonography shows a small fusiform well-defined cystic extramedullary lesion (asterisks) at the dorsal aspect of the thoracic cord (arrows), suspicious of an arachnoid cyst. Corresponding T2-weighted magnetic resonance imaging (c) sagittal and (d) axial images demonstrate the cerebrospinal fluid content and extramedullary location of this lesion (asterisks) at the dorsal aspect of the thoracic cord (arrows), thus confirming the diagnosis.

(Figures 4a and 4b). Magnetic resonance imaging (MRI) is useful for confirming that it contains CSF only and that its location is extra-axial (Figures 4c and 4d). It can also demonstrate the extent of the lesion and any mass effect on the cord.

Syringomyelia

This is a longitudinally oriented CSF-filled cavity within the spinal cord that involves both the central canal and parenchyma.[7] It is frequently associated with Chiari I malformation (41%), tethered cord (25%), trauma (28%), neoplasm (15%), but can also be idiopathic (15%).[7] The lower cervical cord is commonly affected and there can be an extension to the brainstem (syringobulbia).

Ultrasound is useful in its detection, which is characterised by a longitudinally oriented anechoic area within the spinal cord, representing the dilated CSF cavity (Figures 5a to 5c). MRI shows cord enlargement corresponding to a well-defined expanded CSF cavity. There may be T2 hypointense signals within the cavity representing flow voids due to CSF pulsations (Figures 5d and 5e).

Occult Spinal Dysraphism

Fibrolipoma of Filum Terminale

This is a common incidental finding; the reported prevalence in autopsies is 6%.[6] It is usually asymptomatic occasionally causes cord tethering. It can occur in the intra- or extra-dural portions of the filum terminale.

The diagnosis may be missed by ultrasound when the conus medullaris is in its normal position. MRI typically shows a thin linear fat-consistent mass in the filum terminale (Figure 6). MRI is also useful to exclude

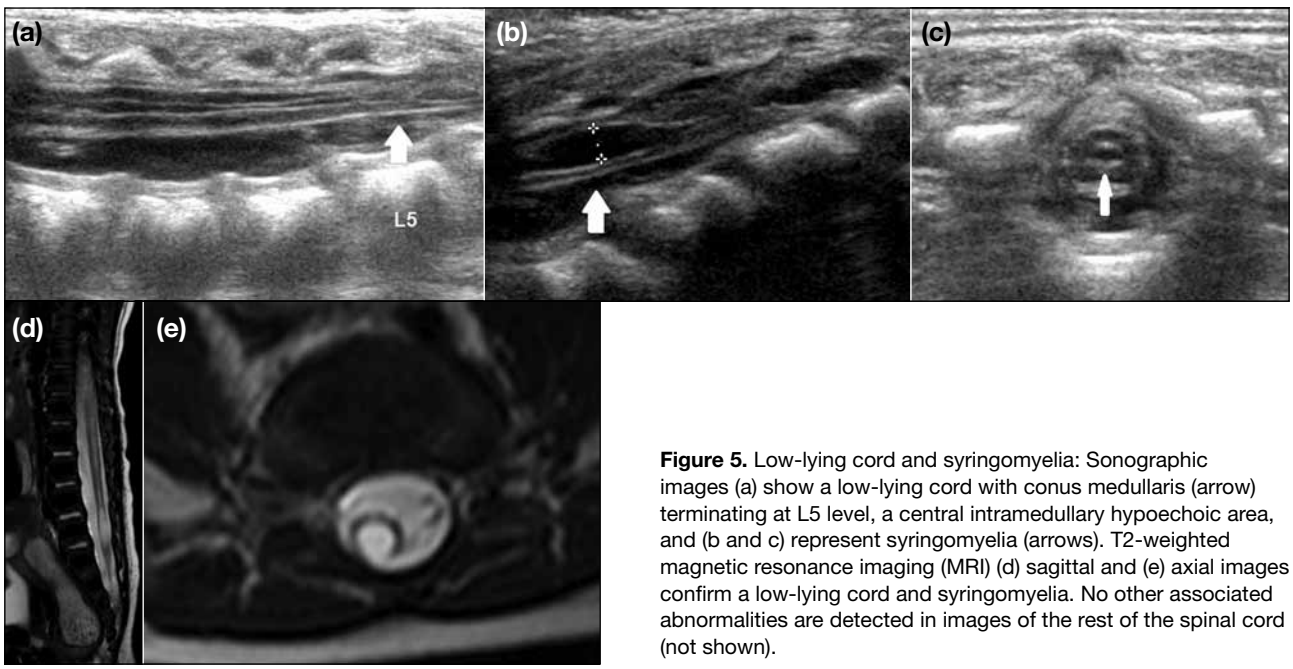

Figure 5. Low-lying cord and syringomyelia: Sonographic images (a) show a low-lying cord with conus medullaris (arrow) terminating at L5 level, a central intramedullary hypoechoic area, and (b and c) represent syringomyelia (arrows). T2-weighted magnetic resonance imaging (MRI) (d) sagittal and (e) axial images confirm a low-lying cord and syringomyelia. No other associated abnormalities are detected in images of the rest of the spinal cord (not shown).

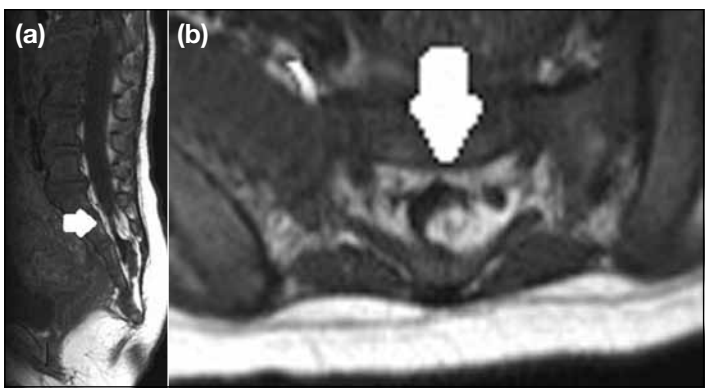

Figure 6. (a) Sagittal and (b) axial T1-weighted magnetic resonance images show a well-defined T1 hyperintense intraspinal mass (arrows) at the S2-4 region, suggestive of a filar lipoma.

associated neural tube anomalies such as tethered cord. The main differential diagnosis is an intradural lipoma. The latter is situated in the subpial region as a juxtamedullary mass, totally enclosed in an intact dural sac.

Myelomeningocele

This is a sac covered by leptomeninges containing CSF and variable amounts of neural tissue, which herniates through a defect in the posterior / anterior elements of spine. It is the most common congenital anomaly of the central nervous system, with an estimated point prevalence of 1 in 1000-2000 births and is twice as common in infants of mothers >35 years old.[7] It is very commonly associated with Chiari II malformations (99%). Other common associations include kyphoscoliosis (90%), hydrocephalus (70-90%), vertebral anomalies, diastematomyelia (20-46%), and chromosomal anomalies (10-17%) such as trisomy 18, trisomy 13, triploidy, and unbalanced translocation.[7] It most commonly affects the lumbosacral area (44%).[7]

Plain films may demonstrate spina bifida and other associated vertebral anomalies. Ultrasound is a good screening tool as it is simple, portable, fast, and non-invasive. Typical ultrasound features include cystic mass in the lumbosacral region with internal linear echogenicities representing neural tissue (Figure 7a). The site of attachment of the neural tissues to the mass is termed the placode. The cord is often low lying (Figure 7b). MRI is often used to further characterise the neural tube defect and screen for other associated neural or skeletal anomalies. Cardinal MRI features include a large cystic mass communicating with the spinal canal through a bony defect with herniation of nerve roots through the defect that is attached to the mass (Figures 7c to 7f).

Surgical repair is the treatment of choice. Mortality from this condition is around 15% by the age of 10 years. Almost all patients have some residual motor dysfunction, whilst 85% achieve social continence through scheduled intermittent catheterization.[7]

Arnold-Chiari Malformation

Chiari II Malformation

This is a disorder of hindbrain dysgenesis, characterised by a caudally displaced 4th ventricle, and brainstem together with tonsillar / vermian herniation through the foramen magnum. It is frequently associated with a lumbar myelomeningocele (>95%) and syringohydromyelia. There may also be associated supratentorial anomalies, such as dysgenesis of the corpus callosum (80-85%), obstructive hydrocephalus (50-98%), absence of septum pellucidum (40%), and stenogyria.[7]

The role of LUS is to identify the myelomeningocele, which is very frequently associated with Chiari II malformations and should prompt further MRI

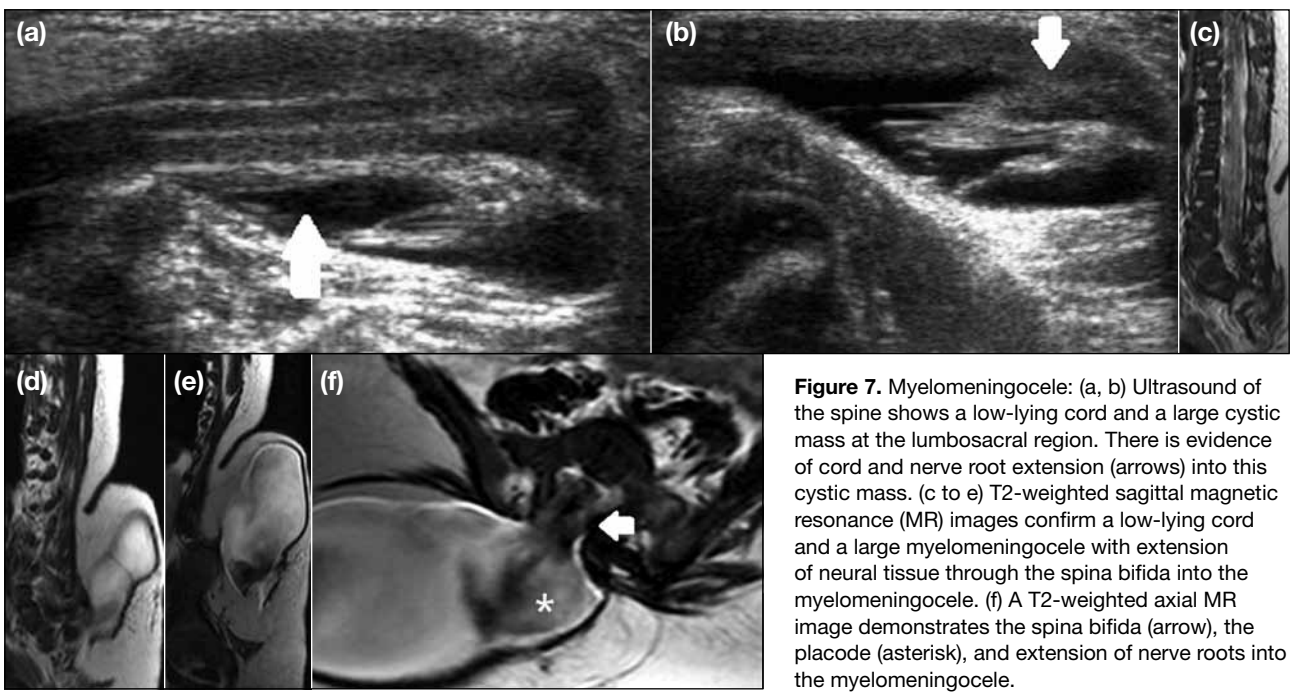

Figure 7. Myelomeningocele: (a, b) Ultrasound of the spine shows a low-lying cord and a large cystic mass at the lumbosacral region. There is evidence of cord and nerve root extension (arrows) into this cystic mass. (c to e) T2-weighted sagittal magnetic resonance (MR) images confirm a low-lying cord and a large myelomeningocele with extension of neural tissue through the spina bifida into the myelomeningocele. (f) A T2-weighted axial MR image demonstrates the spina bifida (arrow), the placode (asterisk), and extension of nerve roots into the myelomeningocele.

investigation of the brain and entire spine to look for cranial features of this malformation. Ultrasound is also helpful in assessing presence of a low-lying cord and syringomyelia, which are other commonly associated anomalies.

MRI of the neuroaxis is the gold standard for the diagnosis and typical features including tonsillar herniation through the foramen magnum into the cervical canal. Hence there is caudal displacement of the 4th ventricle and medulla into the cervical canal with a cervicomedullary kink (Figure 8).

Chiari I Malformation

For the sake of completeness, this malformation is briefly discussed, although by itself it is not an OSD entity. Characteristically there is tonsillar herniation below the foramen magnum but the 4th ventricle and brainstem, though elongated, remain normal in position. It is a frequently isolated anomaly, but can be associated with syringohydromyelia (20-30%), hydrocephalus (25-44%), malformations of the skull base and cervical spine (e.g. basilar impression, craniovertebral fusion, Klippel-Feil anomaly [10%], and platybasia).[7] However, it is not associated with myelomeningocele and therefore sonography of the neonatal spine has little role but may help identify associated syringomyelia. MRI of the neuroaxis is a more appropriate investigation modality that can yield a number of cardinal features. These include downward displacement of cerebellar tonsils and the medial part of the inferior lobes of the cerebellum, which can be more than 5 mm below the level of the foramen magnum (Figure 9). There is inferior pointing (peglike/triangular) of the cerebellar tonsils and obliteration of cisterna magna. The 4th ventricle is elongated but remains normal in position, and there may be slight anterior angulation of lower brainstem.

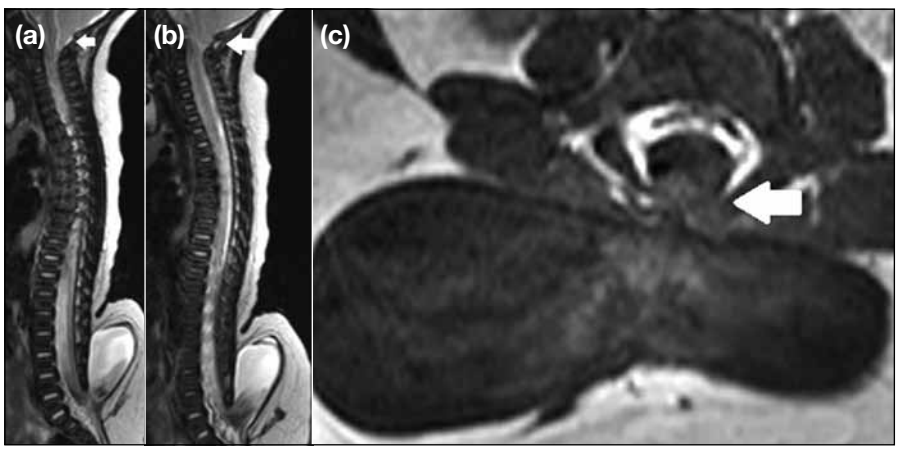

Figure 8. Chiari II malformation: (a, b) T2-weighted sagittal magnetic resonance (MR) images show a lumbosacral myelomeningocele and tonsillar herniation (arrows), suggestive of a Chiari II malformation. (c) A T2-weighted axial MR image demonstrates the spina bifida (arrow) and herniation of neural tissue into the myelomeningocele.

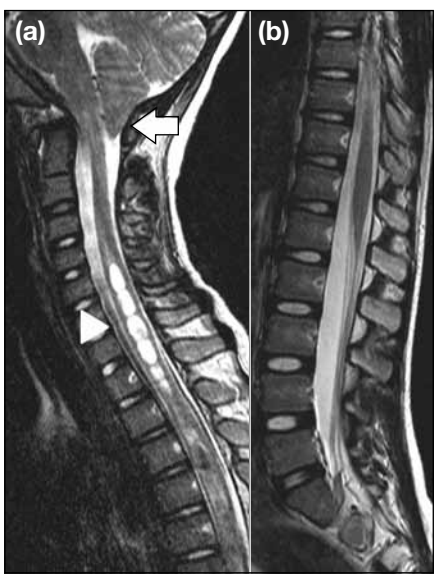

Figure 9. Chiari I malformation: T2-weighted sagittal magnetic resonance images show (a) tonsillar herniation (arrow) and cervical syringomyelia (arrowhead), but (b) a normal position of the conus medullaris.

Sacrococcygeal Teratoma

This is the most common solid congenital tumour in the newborn. It is derived from the residual primitive pluripotent cells during early embryonic development. Notably, 50 to 70% present during first few days of life but may do so up to the age of 2 years, and there is female predominance (4 to 1). It is associated with anomalies in multiple systems, including musculoskeletal (e.g. spinal dysraphism, sacral agenesis, developmental dysplasia of hip), renal (e.g. cystic dysplasia, Potter syndrome), and gastrointestinal (e.g. imperforate anus, gastroschisis).[7]

Altman's classification is commonly used for sacrococcygeal teratomas:

- Type I: predominantly external lesion covered by skin with only a minimal presacral component (47%);
- Type II: predominantly external tumour with a significant presacral component (35%);
- Type III: predominantly a sacral component and external extension (8%); and
- Type IV: a presacral tumour with no external component (10%).

On LUS it can appear solid (25%), mixed (60%), or cystic (15%), with a mean size of 8 cm (Figure 10).[7] If the mass is detected antenatally, associated features of generalised fluid retention indicate a poor prognosis. Such features include polyhydramnios, fetal hydronephrosis, hydrops, and placentomegaly.

MRI is useful for better characterisation and is especially helpful in detecting spinal canal invasion. It typically shows a lobulated and sharply demarcated tumour with a heterogeneous T1 signal due to presence of fat, soft tissue, and calcium. The presence of bony destruction of the sacrum should raise the suspicion of malignancy.

The differential diagnosis is wide ranging and includes common subcutaneous / skin lesions (haemangioma, lymphatic malformation), neural tube anomalies (myelomeningocele, lipomeningocele), bone lesions (chordoma, sarcoma), and rectal lesions (duplication cyst). The presence of fat, soft tissue, and calcification and the absence of neural tissue or communication with the spinal canal are important clues to differentiate teratoma from these conditions. The treatment of sacrococcygeal teratoma entails resection plus coccygectomy.

DISCUSSION

This group of congenital anomalies is referred to as OSD of the spinal canal and its contents, and is characterised by a midline defect affecting the nervous tissue and its bony and soft tissue coverings. By definition, the overlying skin is intact. Frequently OSD is suspected based on prenatal ultrasound, cutaneous stigmata (e.g. sacral dimple, hairy patch, or soft tissue mass) or the presence of congenital anomalies in other body systems. OSD is the most common neonatal spinal malformation, early detection and surgical treatment of which can help to prevent future upper urinary tract problems, infection, and permanent neurological damage.[8-15] Up to 80% of affected patients exhibit a dermal lesion. However, the prevalence of OSD in neonates with only a dermal lesion is reported to be as low as 0.34%, while its prevalence increases dramatically (to 46%) in the presence of cloacal malformations.[12] As a result, the modality of investigation for OSD has to be chosen bearing in mind the risk of the patient population. Radiography, ultrasound and MRI are the major modalities of choice. Radiography has a low specificity (18%) and is not commonly recommended. LUS has a reasonably good sensitivity (87%) and specificity (93%), while MRI has excellent sensitivity (96%) and specificity (91%). Thus, LUS and MRI are the main modalities of choice[16-20]; however the latter is more time-consuming, expensive, and often warrants sedating the patient.

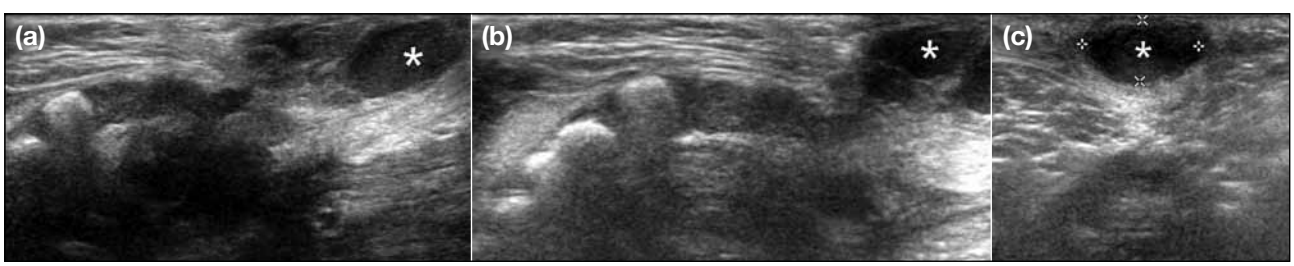

Figure 10. Sonographic images of a 1-day-old baby girl in (a, b) longitudinal and (c) transverse planes show an oval-shaped heterogeneous subcutaneous mass (asterisks) at the sacral region closely associated with the tip of coccyx cranially. Features are compatible with a sacrococcygeal teratoma.

Medina et al,[16] in their cost-effectiveness analysis, found that in low-risk children with an intergluteal dimple, ultrasound was the most effective strategy while in the high-risk group (high anorectal malformation, cloacal malformation, and exstrophy), MRI was actually cost-saving compared with ultrasound. Although an isolated sacral dimple is by far the commonest indication for neonatal LUS, increasing numbers of studies question this practice.[21-24] Robinson et al[13] evaluated 223 infant lumbar spines and found that all 86 patients with an isolated simple (≤5 mm, midline, and <25 mm from the anus) sacral dimple, pit or sinus had no other identifiable abnormality. They therefore proposed an imaging protocol based on the findings of antenatal scans, presence of cutaneous lesions other than a simple dimple, presence of congenital anomalies, and neurological / urological symptoms.[25-28] LUS should only be performed if any of the above was present and isolated simple dimple was no longer an indication.

Chern et al[29,30] also reviewed 943 infants referred for cutaneous stigmata and found seven with OSD. None of the recorded cutaneous stigmata were indicative of OSD and concluded that cutaneous markers were not useful predictors, as the vast majority of LUS studies performed under these circumstances would yield negative findings.

Making the best use of clinical radiology services (6th edition, 2007), published by the Royal College of

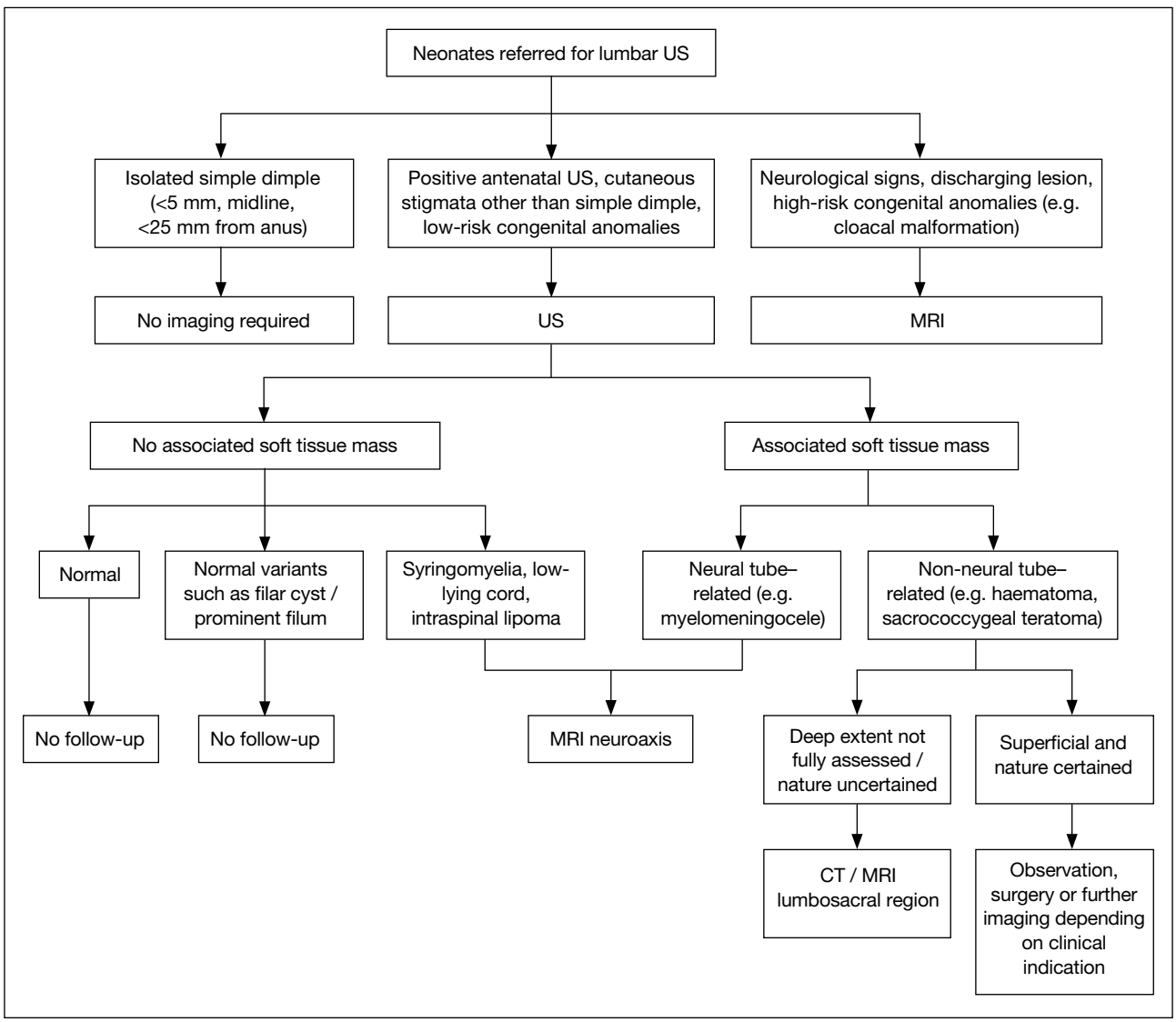

Figure 11. Diagnostic flowchart for neonates referred for lumbar ultrasonography.
Abbreviations: US = ultrasonography; MRI = magnetic resonance imaging; CT = computed tomography.

Radiologists,[17] states that "in the newborn child, isolated sacral dimples and small pits <5 mm from the midline and <25 mm from the anus can be safely ignored. A combination of two or more congenital midline skin lesions is a marker of OSD. If there are other stigmata of spinal dysraphism or associated congenital abnormalities, ultrasound of the neonatal lumbar spine is the investigation of choice. MRI is indicated when ultrasound is abnormal / equivocal, when there are neurological signs, or when there is a discharging lesion".

Based on these, we designed a flowchart summarising the diagnostic strategy for patients referred for neonatal LUS (Figure 11). Given the very low prevalence of OSD in the low-risk population (cutaneous stigmata other than simple sacral dimple) and the reasonably good sensitivity and specificity of ultrasound, it should be the first-line imaging modality in this group of the population. For higher-risk subjects (high anorectal malformation, cloacal malformation, and exstrophy) who have a higher prevalence, ultrasound may not be sensitive enough to rule out an underlying OSD, in which case MRI would be more suitable as the first-line imaging modality.

For patients without an associated soft tissue mass, a negative ultrasound is sufficient for workup and no further follow-up imaging is necessary. Common incidental findings, such as prominent filum or filar cyst, are not associated with OSD and warrant no follow-up imaging. For positive findings such as low-lying cord, intraspinal lipoma, syringomyelia, etc, LUS has a very high specificity in diagnosis, but a further MRI of the entire neuroaxis should still be performed to look for associated intracranial or upper spinal anomalies (such as Chiari I malformation).

In cases with an associated soft tissue mass, ultrasound is useful to delineate its relationship with the spinal canal to determine whether it is related to an underlying OSD. If the mass is related to the spinal canal and a myelomeningocele / meningocele is suspected, MRI should be performed to delineate the anatomy for surgical planning and to evaluate associated CNS anomalies in the brain and upper spine.

For a soft tissue mass not related to the spinal canal, ultrasound may be adequate for diagnosis if the imaging features are characteristic (e.g. lipoma or vascular malformation). If the imaging features are non-specific or the deep extent of the mass is not fully evident, computed tomography or MRI should be performed for further evaluation or surgical planning.

CONCLUSION

The preferred first-line imaging modality for neonatal spine is LUS. It provides adequate screening with high sensitivity and specificity for low-risk subjects presenting with cutaneous stigmata other than simple sacral dimple. It is also useful for diagnosing lumbosacral soft tissue masses that have specific sonographic features. By contrast, MRI is the first-line imaging investigation for persons at high-risk population of OSD (high anorectal malformation, cloacal malformation, and exstrophy). It is useful as a second-line imaging modality for better anatomical delineation of anomalies detected by LUS, which warrant surgical planning as well as screening for associated anomalies in the rest of the neuroaxis.

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