SPINAL DYSRAPHISM: MR IMAGING RATIONALE

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SUMMARY

Spinal cord development occurs through the three consecutive periods of gastrulation (weeks 2-3), primary neurulation (weeks 3-4), and secondary neurulation (weeks 5-6). Spinal cord malformations derive from defects in these early embryonic stages, and are collectively called spinal dysraphisms. Spinal dysraphisms may be categorized clinically into open and closed, based on whether the abnormal nervous tissue is exposed to the environment or covered by skin. Open spinal dysraphisms include myelomeningocele and other rare abnormalities such as myelecule, hemimyelomeningocele, and hemimyelocoele, and are always associated with a Chiari II malformation. Closed spinal dysraphisms are further divided into two subsets based on whether a subcutaneous mass is present in the low back. Closed spinal dysraphisms with mass comprise lipomyelocele, lipomyelomeningocele, meningocoele, and myelocystocele. Closed spinal dysraphisms without mass comprise simple dysraphic states (tight filum terminale, filar and intradural lipomas, persistent terminal ventricle, and dural sinuses) and complex dysraphic states. The latter category involves a developmental abnormal spinal cord development, either in the form of failed midline integration (ranging from complete dorsal enteric fistula to neurenteric cysts and diastematomyelia) or of segmental agenesis (caudal agenesis and spinal segmental dysgenesis). Magnetic resonance imaging is the imaging modality of choice for evaluation of this complex group of disorders.

Key words: spinal cord, dysraphism, MR imaging.

RÉSUMÉ

Dysraphies spinales : aspects enIRM

Le développement de la moelle épinière se déroule au cours de 3 phases successives : la gastrulation (2-3e semaine), la neurulation primitive (3e et 4e semaines) et la neurulation secondaire (5e et 6e semaines). Les malformations de la moelle proviennent de défauts survenus lors de ces phases et sont dénommées dysraphies spinales (vertébro-médullaires). Les dysraphies spinales peuvent être subdivisées en dysraphies fermées ou ouvertes selon que le tissu nerveux anormal est recouvert ou non par la peau. Les dysraphies comprennent les myéloméningocèles et d’autres anomalies rares telles que les myélocèles, les hémomyélocèles et les hémimyélocèles. Ils sont souvent associés à une malformation de Chiar (type II). Les dysraphies fermées sont subdivisées en deux groupes selon que une masse sous-cutanée est présente ou absente à la partie basse du dos. Les dysraphies spinales fermées avec masse regroupent les lipomyélomes, les lipomyélocèles, les lipomyélenoçèles, les méningocèles et les myélomes. Les dysraphies fermées sans masse regroupent les états dysraphiques simples (tîit filum terminale, lipomes intraduraux, persistance de ventricule terminal, sinus dermique) et les états dysraphiques complexes. Cette dernière catégorie correspond à un développement anormal de la notocorde, soit des défauts d’intégration de la ligne médiane (allant de la fistule entérine dorsale complète au kyste neurotérinique et diastématoméyliée) ou l’agenésie segmentaire (agenésie caudale et dysgénésie segmentaire spinales). L’imagerie par résonance magnétique est la méthode de choix pour le diagnostic de ces affections.

Mots-clés : moelle, dysraphie, IRM.

INTRODUCTION

Congenital malformations of the spinal cord are collectively termed spinal dysraphisms. These conditions are usually diagnosed at birth or in early infancy, but some may be discovered in older children or adults. Because of its multiplanar imaging and tissue characterization capabilities, magnetic resonance imaging (MRI) has greatly improved the diagnosis of these disorders and has enhanced the possibility of earlier and case-tailored treatment. Classification is based on a rational correlation of clinical, neuroradiological, and embryological information. Use of classification schemes may prove helpful in making a diagnosis in daily clinical practice [51]. The present paper attempts to summarize the basic concepts about normal and deranged spinal cord embryogenesis, to describe the principal malformations, and to offer a practical approach to neuroradiological decision-making.

EMBRYOLOGY

Spinal dysraphisms result from derangement occurring during a limited period of time during early embryogenesis, i.e., between gestational weeks 2 and 6. The relevant embryogenetic steps are represented by gastrulation (weeks 2-3), primary neurulation (weeks 3-4), and secondary neurulation (weeks 5-6) [50]. During gastrulation, the bilaminar embryonic disk, formed by epiblast (future ectoderm) and hypoblast (future endoderm), is converted into a trilaminar disk with formation of an intervening third layer, the mesoderm. This process begins by day 14 or 15 when a stripe of thickened epiblast composed by totipotent cells, the so-called primitive streak, appears caudally in the midline of the dorsal surface of the embryo. The Hensen’s node is
the knob-like cranial termination of the primitive streak. Epiblastic cells start migrating toward the primitive streak and pass inward at the Hensen’s node to the interface of epiblast and primitive endoderm. Waves of epiblastic cells migrate laterally along the interface form the interposed mesoderm, whereas cells migrating along the midline form the notochord. The notochord, the foundation of the axial skeleton, extends throughout the entire length of the future vertebral column. From the mesoderm surrounding the neural tube and notochord, the skull and vertebral column, and the membranes of the brain and spinal cord are developed.

According to traditional views, the notochord induces the overlying ectoderm to differentiate into neuroectoderm, although recent studies support the hypothesis that the default state of the ectoderm is in fact neural ectoderm [27]. Establishment of the neural plate marks the beginning of primary neurulation. On about day 18, the neural plate starts bending, forming paired neural folds that progressively increase in size and approach each other to eventually fuse in the midline to form the neural tube. According to classical embryological theories, neural tube closure occurs first at the level of the fourth somite (future cranio cervical junction) and proceeds both cephalad and caudad in a zipperlike fashion. The cranial end of the neural tube (rostral neuropore) closes at day 25, whereas the caudal end (caudal neuropore) closes at day 27 or 28. Closure of the posterior neuropore terminates primary neurulation.

The segment of the spine and spinal cord caudad to somite 32 is formed by secondary neurulation, that begins immediately after completion of primary neurulation and proceeds until approximately gestational day 48. During secondary neurulation, an additional part of the neural tube is laid down caudad to the caudal neuropore by the so-called tail bud, a mass of cells deriving from the caudal portion of the primitive streak. The spinal cord formed by secondary neurulation differs from the primary neural tube in many ways. Most notably, while the primary neural tube results from an unfolding of the lateral borders of the neural plate which join at the midline, the secondary neural tube is formed by an infolding of the neural plate, creating an initially solid medullary cord that subsequently becomes cavitated [9, 55]. The secondary neural tube eventually results in the tip of the conus medullaris and the filum terminale, which is a fibroconnectival structure practically devoid of neural elements. This has traditionally been explained through the concept of retrogressive differentiation, in which a combination of regression, degeneration, and further differentiation would occur. However, such process has not been clearly demonstrated in humans, and it is possible that a lack of proliferation, rather than regression or degeneration, contributes to the eventual rudimentary character of the filum terminale (Dr M. Catala, personal communication). A slight expansion of the central canal within the conus medullaris, called terminal ventricle, represents the eventual remnant of the secondary neural tube lumen. It is usually undetectable by MRI in normal individuals.

TERMINOLOGY

Open and closed spinal dysraphisms

The term dysraphism etymologically refers to defective neural tube closure and, therefore, should apply to abnormalities of primary neurulation only. However, it is used to refer to congenital spinal cord disorders in general. Due to the common embryological origin, caudal spinal anomalies are also included in this group.

Spinal dysraphisms are categorized into open spinal dysraphisms (OSD) and closed spinal dysraphisms (CSD) [50] (figure 1). In OSDs, the nervous tissue is exposed to the environment through a congenital bony defect. Conversely, CSDs are covered by skin (i.e., there is no exposed neural tissue), although they are belied by cutaneous birthmarks in as many as 50% of cases [14]. We therefore suggest that the synonymous term “occult spinal dysraphisms” be abandoned, as it implies true, complete absence of tell-tale signs of the underlying abnormality [51].

Spina bifida

The term ‘spina bifida” refers to defective fusion of posterior spinal bony elements [18] but was, and still is, widely used as a synonym of spinal dysraphisms. The terms ‘spina bifida aperta” or “cystica” and “spina bifida occulta” were used in the past to refer to OSD and CSD, respectively [40], but these terms are no longer in widespread use and we do not encourage their utilization.

Placode

The placode is a segment of non-neurulated embryonic neural tissue, i.e., frozen at the neural plate stage. A placode is found in all OSDs as well as in several types of CSDs. It is exposed to the environment in the former, and covered by the integuments in the latter. A placode may be categorized into terminal and segmental depending on location. A terminal placode lies at the caudal end of the spinal cord and may in turn be either apical or parietal, depending on whether the defect involves the apex proper or a longer segment of the cord [51]. A segmental placode lies at an intermediate level along the spinal cord; caudad to the abnormality the cord regains normal morphology and structure [50].

Tethered cord syndrome

Many believe that the tethered cord syndrome (TCS) is some sort of malformation. Instead, it is a clinical syndrome [53, 54] that may ensue as a complication of myelomeningocele repair or as the presentation of several forms of CSD, including spinal lipomas, the tight filum terminale, diastematomyelia, and caudal agenesis. TCS involves traction on a low-lying conus medullaris with progressive neurologic deterioration due to metabolic derangement. The clinical picture includes motor and sensory dysfunction, muscle atrophy, decreased or hyperactive reflexes, urinary incontinence, spastic
SPINAL DYSRAPHISM

Gait, and orthopedic deformities such as scoliosis or foot and hip deformities.

CLASSIFICATION RATIONALE

Classifications of spinal dysraphisms rely on the correlation of radiological features with a specific derangement in the normal developmental cascade. However, traditional classification schemes are continuously challenged by new knowledge emerging about both normal and deranged embryogenesis. From a practical perspective, it is important to use a conceptual framework that relies on identifying those factors that critically restrict the scope of possible diagnoses (table I).

The initial step in this intellectual approach is clinical. Is the malformation exposed to air, or is there intact skin coverage? Simply, the first categorization is between OSD and CSD[50].

OSDs usually cause little, if any, diagnostic puzzle to the neuroradiologist. Only four varieties of OSD exist, with myelomeningocele taking the lion’s share (98.8% of cases)[51]. Clinically, myelomeningoceles are characterized by elevation of the neural placode by the underlying expanded subarachnoid space, whereas in myelocoele the placode is flush with the surface of the back. Such differentiation is clinical and the neuroradiologist plays little part in it. Conversely, it is the neuroradiologist’s role to identify those extremely rare cases in which the closure defects affect one hemi-cord in patients with diastematomyelia. These entities, known as hemimyelocele and hemimyelomeningocele, are very rare. More importantly, the neuroradiologist plays a critical part in the assessment of: the Chiari II malformation; the associated hydrocephalus; and the complications of myelomeningocele repair [20, 24, 42].

CSDs are much more heterogeneous than OSDs. Some are not clinically evident at birth, and patients may elicit clinical attention later in infancy when complications ensue. Clinical examination may help to significantly restrict the focus of the differential diagnosis. A critical factor is the presence of a subcutaneous mass on the patient’s back. In the vast majority of cases, such mass lies at the lumbar or lumbosacral level. Only four malformations will present with a subcutaneous mass in this location, i.e., lipomyelocele, lipomyelomeningocele, meningocele, and terminal myelocystocele [50]. Whereas the latter two are extremely rare, lipomyelocele and lipomyelomeningocele are much more common, and the mass is represented by a subcutaneous lipoma in both cases. In lipomyeloceles, the lipoma gains access of the spinal

Fig. 1. – Clinical features of open and closed spinal dysraphisms. A. Open spinal dysraphism: myelomeningocele. Low back of 1-hour-old newborn prior to surgery. The placode (P) is directly exposed to the environment and is surrounded by partially epithelized skin (membrano-epithelial zone) (asterisk). More laterally, intact skin (S) is elevated by underlying expanded subarachnoid spaces. B. Closed spinal dysraphism with subcutaneous mass. Low back of 1-year-old patient with lipomyelocele. A large subcutaneous mass lies above the intergluteal crease. There is a continuous skin coverage to the abnormality. Notice associated dermal sinus (arrow). C. Closed spinal dysraphism without subcutaneous mass. Back of 2-year-old patient with diastematomyelia. There is a hairy tuft at high lumbar level. Although hairy tufts may belie several varieties of closed spinal dysraphism, diastematomyelia should be suspected when they lie at a relatively cranial level along the child’s back.


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Open spinal dysraphisms

- Myelomeningocele
- Myelocele
- Hemimyelomeningocele
- Hemimyelocele

Closed spinal dysraphisms

With subcutaneous mass: Lumbosacral
- Lipomas with dural defect
  - Lipomyelomeningocele
  - Lipomyelocele
- Terminal myelocystocele
- Meningocele
- Cervico-thoracic
  - Myelocystocele
  - Skin-covered myelomeningocele (limited dorsal myeloschisis)
- Meningocele

Without subcutaneous mass: Simple dysraphic states
- Intradural lipoma
- Filar lipoma
- Cystic filum terminale
- Persistent terminal ventricle
- Dermal sinus

Complex dysraphic states

1. Disorders of midline notochordal integration
   a) Diastematomyelia
   b) Neurenteric cysts
   c) Dorsal enteric fistula

2. Disorders of notochordal formation
   a) Caudal agenesis
   b) Segmental spinal dysgenesis

Open spinal dysraphisms

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Myelomeningocele and Myelocele

Myelomeningocele represents 98.8% of all OSDs [51]. In myelomeningocele, the placoce protrudes together with the meninges through a bony defect in the midline of the back, therefore being exposed to the environment. Because of expansion of the underlying subarachnoid space, the surface of the placoce is elevated above the skin surface. This distinguishes myelomeningocele from the far less common myelocele, or myeloschisis, in which the placoce is flush with the cutaneous surface. As the degree of motor deficit is affected by direct injury to the exposed placoce during delivery, cesarian section is typically performed. Because placoce ulceration and infection are leading causes of mortality in the untreated newborn, affected patients are operated on soon after birth. The subsequent clinical picture includes sensorimotor deficits of the lower extremities, bowel and bladder incontinence, hindbrain dysfunction, intellectual and psychological disturbances. Additionally, it is estimated that 18-40% of individuals with OSDs are allergic to latex. Allergic responses can vary from mild to anaphylaxis, and occur when latex products touch the skin or mucous membranes. Latex-free devices must be used in the MR environment when dealing with these patients.

Embryologically, both myelomeningocele and myeloceles result from faulty primary neurulation [3], with persistence of a portion of non-neurulated placoce. The vast majority of OSDs involve the lumbosacral spine, and the placoce is terminal. However, purely lumbar, thoracolumbar, and thoracic myelomeningocele exist, in which case the placoce is segmental. The exposed surface of the placoce represents the would-be inner surface of the spinal cord and is covered by a rich network of small and friable vessels. The cutaneous ectoderm remains in a lateral position, resulting in a midline skin defect. Because the mesenchyme cannot migrate behind the neural tube, bones, cartilage, muscles and ligaments are forced to develop anterolaterally to the neural tissue, and therefore will appear everted. The ventral surface of the placoce is formed by the would-be external surface of the spinal cord, from which the nerve roots originate. These nerve roots course obliquely through the subarachnoid space to reach their corresponding neural foramina.

Owing to the need for prompt surgical operation, these newborns only very rarely undergo MRI studies prior to surgery. However, we suggest a preoperative MRI investigation should be performed whenever possible, in order to obtain: (1) anatomic characterization of the various components of the
malformation, i.e., the relationships between the placode and nerve roots; (2) presurgical evaluation of the entity and morphology of the malformation sequence (hydromyelia, Chiari II malformation, and associated hydrocephalus) [46]; and (3) identification of rare cases with associated cord splitting (hemimyelomeningoceles and hemimyeloceles). MRI of untreated OSDs shows dehiscence of the subcutaneous fat, fascia, bone, and muscle at level of the spina bifida, and a low position of the spinal cord that forms the dorsal wall of the defect. In myelomeningoceles (figure 2), the subarachnoid spaces are widely dilated and are crossed by nerve roots that arise from the ventral surface of the placode, which is elevated from the cutaneous surface; in myeloceles (figure 3), the placode is flush with the skin.

Hydrocephalus usually appears within 48 to 72 hours after surgery; it is preferentially treated by ventriculoperitoneal shunting, and evaluation of ventricular size often is the major reason for referral to neuroradiological examinations. Other causes for neuroradiological evaluation of these children include subsequent deterioration of a previously stable neurological function. This may be caused by retethering of the spinal cord at the surgical site, (epi)dermoid cysts, or hydromyelia. Retethering by scar is difficult to demonstrate on MRI, as most of the postoperative MRI studies usually show a close relationship between the dorsal surface of the placode and the surgical site, even in patients who do not exhibit TCS. Therefore, scar retethering is usually an exclusion diagnosis. Dysontogenetic masses may result from inadvertent inclusion of epidermal cells during repair of OSDs (inclusion dermoids). They are usually located in close vicinity to the surgical site. They appear as masses that usually are slightly hyperintense to CSF both in T1- and T2-weighted images, and may enhance if superimposed infection is present. Hydromyelia may occur in as many as 80% of operated patients, and may cause rapid development of scoliosis if left untreated [20, 24, 42].

Hemimyelo(meningo)cele

Myelomeningoceles and myeloceles are associated with diastematomyelia in 8-45% of cases [16, 17]. However, if the cord splitting lies at a different level than the placode, the resulting malformation is merely an association between diastematomyelia and OSD. Only when one hemicord fails to neurulate is the malformation called hemimyelocele. When there is associated meningeal expansion, the malformation is called hemimyelomeningocele. If such restrictive definition is accepted, then these anomalies become extremely rare [51]. Clinically, neurological impairment is similar to that seen in patients with diastematomyelia, but it is markedly asymmetric [35]. The presence of hirsutism along one side of the exposed neural defect may be strongly suggestive of underlying cord splitting.

Embryologically, these malformations are related to faulty gastrulation (see section on diastematomyelia), with superimposed failure of primary neurulation of one hemicord.

Chiari II malformation

There is a 100% association between OSDs and the Chiari II malformation, a congenital hindbrain
anomaly characterized by a small posterior fossa with caudal displacement of the vermis, brainstem, and fourth ventricle (figure 2) [6]. McLone and Knepper [25] proposed a theory to explain this consistent association. Normally, the medial walls of the primitive central canal of the neural tube (“neurocele”) appose and occlude the neurocele transiently during primary neurulation. Failure to occlude the neurocele allows free downward CSF flow. Therefore, CSF leaks freely through the spinal defect into the amniotic sac because the neural tube remains non-neurulated. This results in chronic CSF hypotension within the developing neural tube. Consequently, the rhombencephalic vesicle fails to expand, causing lack of induction of the perineural mesenchyme of the posterior cranial fossa. Both the cerebellum and brain stem eventually are forced to develop within a smaller than normal posterior fossa and consequently herniate through both the tentorial groove and the foramen magnum [29].

CLOSED SPINAL DYSRAPHISMS

CSDs with subcutaneous mass

These abnormalities are characterized by a skin-covered mass belying the underlying malformation. Most often, the mass lies at the lumbosacral level right above the intergluteal crease, and the corresponding anomalies are represented by four entities: the quite common lipomas with dural defect (lipomyelocèle and lipomyelomeningocele), and the distinctly uncommon terminal myelocystocele and meningocele [50]. Differential diagnosis basically includes sacrococcygeal teratomas, whose location is more caudal, i.e., at or below the intergluteal crease. Cervico-thoracic CSDs with associated subcutaneous masses are distinctly uncommon.

Lipomas with dural defect: lipomyelocèle and lipomyelomeningocele

Lipomyelmetocèles and lipomyelomeningoceles are characterized by a midline subcutaneous fatty mass right above the intergluteal crease, usually extending asymmetrically into one buttock [30]. Because the mass is clinically evident at birth, the diagnosis usually is obtained before neurological deterioration ensues. Infants who are not treated before 6 months of age usually develop hypostenia and hypotrophy of muscles of both lower limbs, gait disturbances, urinary incontinence, and paresthesias. These clinical features progress over time if the child is left untreated.

Histologically, the mass is composed by clusters of mature adipocytes separated by collagenous bands, usually associated with other tissues such as striated muscle, cartilage, bone, nerve cells, ependyma, and aberrant neuroglial tissue. Sometimes, these aberrant tissues coalesce to form hamartomatous masses that have been designated “dyssympathic hamartomas”. Although congenital intraspinal lipomas are anatomically stable [26], they may grow as part of the normal increase of adipose tissue throughout childhood [24], other than in particular conditions such as obesity or pregnancy.

Embryologically, spinal lipomas are abnormalities of primary neurulation. They are traditionally believed to result from focal premature disjunction of the cutaneous ectoderm from the neuroectoderm, allowing mesenchyme to access the interior of the neural tube, where it will be induced to form fat by contacting the ependymal lining [27, 30]. Other embryological mechanisms have been invoked, involving abnormalities of the dorsal mesoderm that could either be primitive or secondary to defective induction from the neural tube [8].

The anatomic position of the connection between the spinal cord and the lipoma (i.e., the placode-lipoma interface) varies depending on the size of the lipoma and the degree of expansion of the subarachnoid spaces. In lipomyelocèle (synonym: lipomyeloschisis) (figure 4), the placode-lipoma interface lies within or at the edge of the spinal canal. Both the bony defect and the subcutaneous fat that extend into the spinal canal and attach to the cord are clearly demonstrated by MRI. The placode-lipoma interface may extend over several vertebral levels. It may be smooth and regular, or large and irregular, with stripes of fat permeating the exterior of the spinal cord and even penetrating into the central canal. Hydromyelia usually is present in these cases. The size of the spinal canal may be increased depending on the size of the lipoma, but the size of the subarachnoid space ventral to the cord is consistently normal.

Lipomyelomeningoceles may produce a constellation of MRI features. Individual cases typically differ from one another, depending on the relative size of the meningocele and lipoma as well as the orientation of the placode. However, the placode-lipoma
The placode-lipoma interface lies outside the anatomic boundaries of the spinal canal in all cases. An archetypal condition in which the placode-lipoma interface lies exactly along the midline within the meningocele is not the rule but, rather, an exception [50] (figure 5). In most instances, the placode is stretched and rotated eccentrically towards the lipoma on one side, whereas the meninges herniate on the opposite side (figure 6). In such an instance, the spinal roots emerging from the side facing the meningocele are generally longer and may be at risk for surgical trauma, whereas those lying on the side of the lipoma are shorter and cause spinal cord tethering. Different from lipomyeloceles, the spinal canal is dilated due to expansion of the subarachnoid spaces ventral to the cord.

**Meningocele**

The classic posterior meningocele results from herniation of a CSF-filled sac lined by dura and arachnoid through a posterior bony spina bifida (figure 7). It is commonly lumbar or sacral in location, but thoracic and even cervical meningoceles may be found. In general, spinal meningoceles are less common than is usually believed (2.4% of all CSDs) [1]. Their embryogenetic origin is unknown, but they might result from ballooning of the meninges through a posterior spina bifida due to the relentless effect of CSF pulsations. Although both nerve roots and, more rarely, a hypertrophied filum terminale may course within the meningocele, by definition no part of the spinal cord enters the sac. The spinal cord itself is completely normal structurally, although it usually is tethered to the neck of sacral meningoceles.

Anterior meningoceles almost always are presacral in location and are consistently found within the setting of caudal agenesis [23]. They are usually discovered in older children or even in adults when MRI is performed for low back pain, urinary incontinence, or constipation.

**Terminal myelocystocele**

Terminal myelocystoceles involve expansion of the central canal of the caudal spinal cord produced by terminal hydromyelia (“syringocele”), surrounded by an expanded dural sheath (“meningocele”). A subcutaneous lipoma is usually associated, so that the abnormality could perhaps be better defined as a “lipomyelocystocele”. The inner terminal cyst communicates with the central canal of the spinal cord (figure 8), whereas the outer dural sac communicates with the subarachnoid space. The outer and inner fluid spaces usually do not communicate [5, 26, 36]. Patients with terminal myelocystoceles typically have no bowel or bladder control and poor lower-extremity function [6]. The prognosis is mainly related to associated anomalies, usually belonging to the OEIS constellation (omphalocele, exstrophy of the cloaca, imperforate anus, spinal anomalies) [30, 31].

The embryological origin of the terminal myelocystocele has not yet been determined. Current theories postulate a perturbation of CSF dynamics [5, 26, 36]. Inability of CSF to exit from the early neural tube could cause the terminal ventricle to balloon into a cyst that disrupts the overlying mesenchyme. Therefore, the terminal myelocystocele could be viewed as a severe, disruptive variety of a persistent terminal ventricle [50].
Fig. 5. – Lipomyelomeningocele, 1-year-old boy. A. Sagittal T1-weighted image shows large subcutaneous lipoma. The spinal cord exits the spinal canal through a posterior sacral spina bifida and ends into a terminal apical placode (P) that connects to the inner surface of the lipoma (black arrowheads). Notice concurrent hydromyelia (white arrows). B. Axial T1-weighted image shows terminal apical placode. Expansion of the subarachnoid spaces causes the placode to bulge outside the anatomic boundaries of the spinal canal through a wide posterior spina bifida (white arrows). The meningocele (M) develops symmetrically to both sides of the placode (P). The placode-lipoma interface lies on the midline (black arrowheads).

Fig. 5. – Lipomyéloméningocèle, enfant âgé d’un an. A. Séquence pondérée en T1 – coupe sagittale : lipome sous-cutané. La moelle sort du canal à travers une spina bifida sacrée postérieure et se termine dans une placode apicale terminale (P) qui est reliée à la partie interne du lipome (tête de flèche blanche). Noter l’hydromyélie (flèche blanche). B. Séquence pondérée en T1 – coupe axiale : placode apicale terminale. Extension des espaces sous-arachnoïdiens responsables d’un bombement de la placode au-delà des limites anatomiques du canal rachidien à travers un large spina bifida postérieur (flèche blanche). La méninocèle (M) se développe de façon symétrique des deux côtés de la placode (P), la limite entre lipome et placode s’étant sur la ligne médiane (tête de flèche noire).

Fig. 6. – Lipomyelomeningocele, 40-day-old girl. A. Sagittal T1-weighted image shows large meningocele (M) contained within a huge subcutaneous lipoma. The spinal cord (white arrow) is seen to approach the neck of the meningocele in this midsagittal view. Notice the last visible vertebra is S2, consistent with an associated picture of caudal agenesis. B. Axial T1-weighted image shows the spinal cord projects out of the spinal canal and courses along the left side of the meningocele (M) to connect to the lipoma (arrowhead); therefore, the placode (P) is terminal apical and the off-midline placode-lipoma interface lies outside the anatomic boundaries of the spinal canal, consistent with a diagnosis of lipomyelomeningocele.

Fig. 6. – Lipomyéloméningocèle, petite fille de 40 jours. A. Séquence pondérée en T1 – coupe sagittale. Large méningocèle (M) avec lipome sous-cutané. La moelle (flèche blanche) est proche du collet de la méningocèle sur la vue sagittale médiane. Noter que la dernière vertèbre visible est S2, probable traduction d’une agénésie caudale associée). B. Séquence pondérée en T1 – coupe axiale. La moelle est en dehors du canal rachidien et est située à la partie gauche de la méningocèle (M) pour rejoindre le lipome (tête de flèche) : la placode (P) est termino-apicale et l’interface placode – lipome est située en dehors des limites anatomiques du canal rachidien ce qui évoque le diagnostic de lipomyéloméningocèle.
CSDs without subcutaneous mass
Simple dysraphic states

This subset of abnormalities is heterogeneous from an embryological perspective, as it involves defects of both primary and secondary neurulation.

However, they may be grouped from a clinical standpoint, as they represent the most common abnormalities found in relatively older children who usually do not have significant low-back cutaneous stigmata but complain with TCS [45, 47].

Intradural and intramedullary lipoma

Intradural and intramedullary lipomas do not differ from lipomas with dural defects both in pathological and embryological terms. However, they are contained within an intact dural sac. Intradural lipomas lie along the midline in the groove formed by the dorsal surface of the unapposed folds of the placode, and may bulge posteriorly in the subarachnoid spaces elevating the pia mater. Large lipomas may displace the cord laterally, resulting in an off-midline placode-lipoma interface. In rare instances, lipomas are completely intramedullary. Intradural lipomas are commonly located at lumbosacral level (figure 9) and usually present with TSC, whereas cervicothoracic lipomas generally produce insidious signs of spinal cord compression (figure 10). At MRI, lipomas appear as masses that are isointense to subcutaneous fat on all sequences, including those acquired with fat suppression (figure 10) [50].

Filar lipoma

Filar lipoma is an elementary anomaly of secondary neurulation characterized by fibrolipomatous thickening of the filum terminale. The occurrence of incidental fat within the filum terminale in the normal adult population is estimated to be 1.5 to 5% of unselected MRI studies [4, 52]. Therefore, it may be considered an anatomic variant if there are no signs of TCS. The exact embryo-
logical mechanisms by which lipomas of the filum arise remain unknown, but impaired canalization of the tail bud and persistence of cells capable of maturing into adipocytes are likely to be involved [52]. MRI detects fatty tissue within a thickened filum terminale as a stripe of increased signal intensity on sagittal T1-weighted images (figure 11). Because the filum frequently lies slightly off the midline, axial and coronal T1-weighted images are useful to make the diagnosis.

**Fig. 9.** – Intradural lipoma, 2-month-old girl. A. Sagittal T1-weighted image shows low-lying spinal cord tethered (small arrowheads) to the anterior surface of a lumbosacral lipoma (L). The lipoma is not continuous with the subcutaneous fat. Notice concurrent dermal sinus (large arrowhead). B. Axial T1-weighted image shows the placode (P) -lipoma (L) interface (arrowheads). The lipoma is intradural and clearly separated from the subcutaneous fat.

**Fig. 9.** – Lipome intradural, fille de 2 mois. A. Séquence pondérée en T1 – coupe sagittale. Moelle attachée à la face antérieure d’un lipome lombosacré (L). Le lipome n’est pas en continuité avec la graisse sous-cutanée. Noter l’existence d’un sinus dermique (flèche large). B. Séquence pondérée en T1 – coupe axiale. Interface lipome (L) – placode (P) (tête de flèche). Le lipome est intradural est séparé de la graisse sous-cutanée.

**Fig. 10.** – Intradural cervical lipoma. A. Sagittal T1-weighted image show huge intradural lipoma that fills the spinal canal almost completely. The spinal cord is compressed. The spinal canal is enlarged, with scalloped posterior vertebral walls. B. Fat-suppressed sagittal T1-weighted image confirms the fatty composition of the mass. Courtesy Prof. U. Aydingoz, Hacettepe, University, Turkey.

**Fig. 10.** – Lipome cervical intradural. A. Séquence pondérée en T1 – Coupe sagittale. Large lipome intradural occupant la quasi-totalité du canal rachidien. La moelle est comprimée et le canal élargi avec scalping postérieur des corps vertébraux. B. Séquence pondérée en T1 avec suppression de graisse. Coupe sagittale. Confirmation du caractère graisseux de la masse. Clichés dus à l’obligence. Courtesy Prof. U. Aydingoz, Hacettepe, University, Turkey.
Tight filum terminale

The tight filum terminale is characterized by a short, hypertrophic filum terminale that produces tethering and impaired ascent of the conus medullaris. Isolated cases are extremely uncommon, whereas the abnormality is more frequent in patients with other malformations, such as diastematomyelia (figure 12) or dermal sinuses [50]. A low-lying conus medullaris is frequently, albeit not necessarily, associated. In 86% of patients, the tip of the conus medullaris lies inferior to L2 [55].

Embryologically, the tight filum terminale has been related to abnormal retrogressive differentiation of the secondary neural tube, producing a thicker than normal filum. The filum terminale is not
>2 mm in diameter in normal individuals [57], but the exact thickness of the filum terminale may be difficult to measure at MRI.

**Dermal sinus**

The dermal sinus is an epithelium-lined fistula that extends inward from the skin surface and can connect with the CNS and its meningeal coating. It is found more frequently in the lumbosacral region, although cervical, thoracic, and occipital locations are possible [1]. On clinical examination, a midline dimple or pinpoint ostium is found (figure 1), often in association with hairy nevus, capillary hemangioma, or hyperpigmented patches. The cutaneous opening of a dermal sinus tract differs from that of a sacrococcygeal fistula. Dermal sinus tracts are found above the intergluteal cleft and usually are directed superiorly. By comparison, sacrococcygeal pits are found within the natal cleft extending either straight down or inferiorly. They are anatomically located below the level of the cul-de-sac of the subarachnoid space and do not require further imaging evaluation [56]. Although the cutaneous abnormality usually is evident at birth, some patients are not referred to medical attention until they develop complications such as local infection or meningitis and abscesses that may result from bacteria invading the CNS along the dermal sinus tract.

Embryologically, dermal sinus tracts are traditionally believed to result from focal incomplete disjunction of the neuroectoderm from the cutaneous ectoderm [16]. Dermal sinuses are easily recognized on midsagittal MR images as a thin hypointense stripe within the subcutaneous fat (figure 9 and 13), whereas they are more difficult to detect on axial scans. The intrathecal portion of the tract usually is not detectable on MRI, which makes it difficult to assess the true extent of the tract itself and, particularly, whether it pierces the dura and involves the CNS. In a considerable percentage of cases, dermal sinuses are associated with dermoids, generally located at level of the cauda equina or near the conus medullaris and probably resulting from encystment of part of the dermal sinus.
tract (figure 14). This association was found in 11.3% of cases in our series [51], but may be higher. Dermoids show variable MRI features depending on their content. Some portions may be hyperintense on T1-weighted images, but the mass may be isointense to CSF on both T1- and T2-weighted images and, therefore, may be difficult to discern. Infected dermoids exhibit intense contrast enhancement that may be ring-like if an abscess develops.

**Persistent terminal ventricle**

The “fifth ventricle” of the historic scientific literature [21] is a small ependyma-lined cavity within the conus medullaris that always is identifiable on post-mortem examinations but must achieve a certain size to be visible at MRI (figure 15) [10]. Embryologically, it is related to incomplete regression of the terminal ventricle during secondary neurulation, with preservation of its continuity with the central canal of the rostral spinal cord. The latter point is critical because failure of regression of the terminal ventricle associated with inefficient connection to the central canal above may produce a terminal myelocystocele, which is a much more severe abnormality.

By itself, the persistent terminal ventricle is asymptomatic; however, cases have been reported in which a huge cystic dilation was associated with low-back pain, sciatica, and bladder disorders [44]. It is unclear whether these “terminal ventricle cysts” are developmental variants or result from pathological changes leading to obstruction of the terminal ventricle [10]. Differentiation with hydromyelia is based on the location immediately above the filum terminale, whereas intramedullary tumors are excluded by the lack of gadolinium enhancement. The size of the “cyst” usually remains unchanged on follow-up scans.

**Complex dysraphic states**

Because gastrulation is characterized by the development of the notochord, spinal dysraphisms originating during this period will characteristically show a complex picture in which not only the spinal cord, but also other organs deriving from or induced by the notochord, are severely abnormal. Therefore, disorders of gastrulation are also called complex dysraphic states [13]. In the vast majority of cases, these abnormalities are covered by skin, and no tell-tale subcutaneous masses are present. The only exceptions are hemimyelocele and hemimyelomeningocele, two exceedingly rare abnormalities that were described in the “open spinal dysraphism” section.

Failures of notochordal development have been categorized in two subsets: (1) **disorders of midline notochordal integration**, which result in longitudinal splitting, and (2) **disorders of notochordal formation**, which result in the absence of a certain notochordal segment [50]. A thorough discussion of all complex dysraphic states is beyond the scope of this paper. Only the main entities will be dealt with here.

**Disorders of midline notochordal integration**

Embryologically, prospective notochordal cells are derived from the Hensen’s node, and stream in equal numbers from both sides of the node past the primitive pit to migrate between the ectoderm and the endoderm in the midline. Midline integration [51] is the process by which the two paired notochordal anlagen fuse in the midline to form a single notochordal process. The cause of failed midline notochordal integration has been the source of continuing debate among authors, and several possible explanations have been proposed [17]. The eventual malformation depends on the severity of the abnormality and the outcome of the repair.
efforts of the embryo. Several malformations belong to this wide group [51]. Only the most important entities will be dealt with here.

Neurenteric cysts

The most severe form of failed midline notochordal integration is dorsal enteric fistula, in which an abnormal canal connecting the skin surface with the bowel crosses the intervening space between a duplicated spine [51]. Complete dorsal enteric fistula has only exceptionally been reported in the literature. Neurenteric cysts, usually located anterior to the spinal cord in close connection with vertebral abnormalities, probably represent localized forms of dorsal enteric fistula. They are lined with a mucin-secreting, cuboidal or columnar epithelium that resembles the alimentary tract [51]. Their content is variable, and the chemical composition may be similar to CSF in some cases. The typical location is intradural in the cervicothoracic spine anterior to the cord (figure 16); however, neurenteric cysts also may be found in the lumbar spine and even in the posterior fossa.

Embryologically, neurenteric cysts are related to endodermal differentiation of primitive streak remnants that remain trapped between a split notochord. Owing to the common underlying embryological mechanism, neurenteric cysts frequently are associated with diastematomyelia, in which case they may be located in the cleft between the two hemicords.

On MRI (figure 16), neurenteric cysts usually are isointense to hyperintense relative to CSF on long relaxation time sequences. On T1-weighted images, they appear isointense or slightly hyperintense to CSF, consistent with high-protein content. Absence of contrast enhancement is the rule; however, we have seen one case of a neurenteric cyst that enhanced following intravenous gadolinium administration [51].

Diastematomyelia

The most frequent form of failed midline integration of the notochord is diastematomyelia. Diastematomyelia (literally, split cord) refers to a variably elongated separation of the spinal cord in two usually symmetric halves. Whether this malformation represents true cord splitting or cord duplication has been the subject of enduring debate. In fact, there is a continuous spectrum of abnormality ranging between partially cleft cord in a single dural tube at one end and completely duplicated cord within dual dural tubes with intervening bony spur at the other end. The term “split cord malformations” (SCM) has been suggested to describe this malformative continuum [33, 35]. We believe this is just an English translation of the original Greek derived term, diastematomyelia. We therefore encourage to retain the traditional denomination, which also has the advantage of being widely recognized and accepted in the literature.

Abnormal midline integration results in two paired notochordal processes separated by intervening primitive streak cells. Each heminotochord induces a separate neural plate. The resulting malformation depends on the developmental fate of the intervening primitive streak tissue. If it further develops toward bone and cartilage, the two hemicords eventually will be contained into two individual dural sacs separated by an osteocartilaginous spur. Conversely, if the primitive streak tissue is reabsorbed or leaves a thin fibrous septum, the two hemicords eventually will lie within a single dural tube. This represents the foundation of classification of diastematomyelia into two groups [33].

Diastematomyelia type 1 consists of two hemicords contained within individual dural tubes, separated by a bone or osteocartilaginous septum that extends from the vertebral body to the neural arches [33]. This rigid median septum is entirely extradural. Clinically, patients usually present with scoliosis and TCS. A hairy tuft lying high along the child’s back is a very reliable clinical marker of diastematomyelia [1]. Vertebral anomalies are the rule and include bifid lamina, widened interpediculate distance, hemivertebrae, bifid vertebral, fused vertebrae, and narrowing of the intervertebral disk space. Scoliosis also is common and is seen in 30-60% of these individuals.

The radiological hallmark [28] is the osseous or osteocartilaginous septum with resulting double dural tubes, each containing a hemicord (figure 17). Although in the archetypal case the spur is osseous and connects the vertebral body to the neural arch along a midsagittal plane, “atypical” spurs are common. The spur may course obliquely, and may be complete or incomplete, in which case it may originate either from the vertebral body or from the neural arch. In some cases, it divides the spinal
Spinal Dysraphism

Canal unequally, and the two hemicords will be asymmetric. In young children, the spur may be mostly cartilaginous and progressively ossifies as the child grows. In most cases of diastematomyelia type I, the cleft is located at the thoracic or lumbar level and lies at the caudal end of the cord splitting. The two hemicords usually surround the spur tightly before fusing with each other to form a normal spinal cord below, whereas rostrally the splitting is much more elongated. Therefore, there is a cranio-caudal sequence of partial clefting, complete diastematomyelia with single dural tube, and diastematomyelia with dual dural tubes (figure 18).

Hydromyelia is a common associated finding and may involve the normal cord both above and below the splitting, as well as one or both hemicords (figure 18) [41]. In the vast majority of cases, the hemicords fuse again below the spur to form a normal cord. In rare cases, the cleft may be terminal, in which case two hemiices and hemifilum (generally hypertrophic and tethered) are present. Failure of neurulation of one hemicord produces a hemimyocele or hemimyelocele.

Diastematomyelia type II has a common dural tube housing both hemicords without intervening rigid median septum [33, 28]. Three variants exist: presence of an intervening fibrous septum, absence of a septum, and partial cord splitting [50].

A midline, nonrigid, fibrous septum sometimes is detected at surgery. In these cases, clinical signs of TCS may appear, and indeed the assumption that diastematomyelia type II does not produce TCS is incorrect [35]. These septa may be identified on axial and coronal T2-weighted images as thin hypointense stripes interposed between the two hemicords (figure 19). Absence of a septum is the most common occurrence in diastematomyelia type II (figure 20). Although the diagnosis is relatively straightforward both on axial and coronal MR images, diastematomyelia type II may be difficult to appreciate on sagittal MR images, where the only tell-tale sign is an apparent thinning of the spinal cord that results from partial averaging with the intervening subarachnoid space between the two hemicords (figure 19 and 20) [50]. In rare cases, the cleft is partial and the splitting incomplete; these are the mildest forms of diastematomyelia (figure 21) [49].

Hydromyelia may be present with the same features as in diastematomyelia type I. The conus medullaris is typically low, and there is a strong association with tight filum terminale and filar lipomas. Associated vertebral anomalies are usually milder than in type I, and are represented by butterfly vertebrae in most cases. Posterior spine bifida is often present, whereas scoliosis is usually absent.

Disorders of notochordal formation

Programmed cell death, or apoptosis, is a process of cell elimination that occurs during normal development and represents a crucial phenomenon in various steps of embryogenesis. During abnormal gastrulation, prospective notochordal cells that are wrongly specified in terms of their rostrocaudal positional encoding are eliminated. Eventually, fewer cells or even no cell will form the notochord at a given abnormal segmental level. The consequences of such a segmental notochordal paucity are manifold and affect the development of the spinal column and spinal cord as well as other organs that rely on the notochord as their inductor. If the prospective notochord is depleted, a wide array of segmental vertebral malformations...
FIG. 18. – Diastematomyelia type I with hydromyelia, 1-year-old girl. A. Axial CT scan shows obliquely coursing bony spur (S), separating the abnormally large spinal canal into two asymmetric parts. There is associated left posterior spina bifida. B. Parasagittal, C. mid-sagittal, and D. coronal T1-weighted images shows asymmetric bony spur (S) containing high-signal bone marrow and projecting into the spinal canal. The spur is located at the bottom end of the cord splitting, and each hemicord contains hydromyelic cavities (arrowheads). There also is hydromyelia involving the spinal cord both above and below the splitting (arrows). Multiple vertebral segmentation defects involving the lumbar spine and sacrum are revealed by rudimentary intervertebral disks. E-G. Axial T2-weighted images show the malformation sequence from cephalad to caudal: E. hydromyelia (asterisks) with initial cord separation, F. split spinal cord (hc) within single dural sac associated with right hydromyelia (asterisk), and G. split spinal cord with dual dural sacs and intervening bony spur (S). Axial views also show the right hemicord has one paramedian set of nerve roots connected to the bony spur (thin arrow, G); anterior and posterior set of nerve roots is also seen (arrowheads, G).


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including segmentation defects, indeterminate or block vertebrae, or absence of several vertebrae, will result. Because of lack of neural induction and absence of a floor plate, fewer prospective neuroectodermal cells, or even no cell at all, will be induced to form the neural tube in the pathological segment [48]. The resulting malformation essentially depends on the segmental level and the extent of the abnormality along the longitudinal embryonic axis, with subsequent interference on the processes of primary and/or secondary neurulation. In the vast majority of cases, the abnormality involves the caudal extremity of the embryo, resulting in the caudal agenesis constellation. Much more rarely, the abnormality involves an intermediate notochordal segment, thereby resulting in segmental spinal dysgenesis [48].

Caudal agenesis

Caudal agenesis (CA) is a heterogeneous constellation of anomalies comprising total or partial agenesis of the spinal column, anal imperforation, genital anomalies, bilateral renal dysplasia or aplasia, pulmonary hypoplasia, and lower limb abnormalities [15]. CA is also commonly, albeit inappropriately, called caudal regression syndrome; etymologically, the term “caudal agenesis” should be preferred, as “caudal regression” implies a concept of excessive regression of the embryonic tail that cannot be adequately applied in tail-less animals, such as humans (M. Catala, personal communication). Agenesis of the sacrococcygeal spine may be part of syndromic complexes such as OEIS (omphalocele, cloacal exstrophy, imperforate anus, and spinal deformities) [7], VACTERL (vertebral abnormality, anal imperforation, cardiac anomalies, tracheoesophageal fistula, renal abnormalities, limb deformities) [45], and the Currarino triad (partial sacral agenesis, anorectal malformation, and presacral mass: teratoma and/or meningocele) [11, 12, 19]. There is a definite association with maternal diabetes mellitus (1% of offspring of diabetic mothers). CA in humans can be inherited as an autosomal dominant condition [9].

The congenital spectrum of vertebral abnormality in CA may range from agenesis of the coccyx to absence of the sacral, lumbar, and lower thoracic vertebrae, but the vast majority of these anomalies
involve only the sacrum and coccyx. The sacrum may be totally or subtotally absent, with S1 through S4 present in individual cases. Sacral aplasia may be asymmetric, with resulting total or subtotal hemisacrum that may, in turn, be unilateral or bilateral [34]. The heterogeneous spectrum of vertebral malformation requires anteroposterior and lateral plain radiographs for full appreciation. These radiographic studies constitute an essential part of the neuroradiological workup.

Traditionally, CA has been categorized into two types depending on the location and shape of the

![Image](image_url)
conus medullaris: either high and abrupt (type I) or low and tethered (type II). Although these two types were believed to be embryologically related to disordered primary or secondary neurulation, respectively [32], both are actually consistent with an earlier abnormality of gastrulation [50]. Segmental maldevelopment of the caudal notochord and axial-paraxial mesoderm results in an abnormality that interferes with either secondary neurulation alone, or both primary and secondary neurulation, depending on the longitudinal extent of the original notochordal damage [50]. Therefore, the crucial embryological watershed between the two varieties is the interface between primary and secondary neurulation (i.e., the junction between the true notochord and the tail bud), corresponding to the caudal end of the future neural plate. This site has been the source of continuing debate among authors: recent data suggest that it corresponds to S3 through S5 [31]. Based on this theory, the degree of spinal cord aplasia correlates with the severity of the spinal malformation, with a greater degree of vertebral aplasia in type I than in type II [32].

**Type I CA** (figure 22 and 23). If not only the tail bud, but also part of the true notochord fails to develop, interference is generated with both the processes of primary and secondary neurulation [50]. Depending on the severity of the original damage, the eventual degree of vertebral aplasia will range from absence of the coccyx and lower midsacrum to aplasia of all coccygeal, sacral, lumbar, and lower thoracic vertebrae (figure 22). However, the last vertebra is L5 through S2 in the majority of patients. Owing to the same embryological mechanism, there is aplasia of the caudal metameres of the spinal cord. This results in an abrupt spinal cord terminus that nearly always is club or wedge shaped (figure 23) [2]. The spinal cord terminus is high-lying (most often opposite T12) in most cases, but it may lie opposite to L1 in a minority of cases (figure 22). The cauda equina also is frequently abnormal, and the nerve roots have an abnormal course that has been termed the “double bundle shape” [34]. The thecal sac tapers below the cord terminus, and ends at an unusually high level (figure 23). Associated caudal anomalies, such as anterior meningoceles and teratomas, may be present, although much less frequently than in type II CA. Unlike in type II, the cord is not tethered to these caudal anomalies. This accounts for the negligible proportion of TCS with progressive neurological deterioration in these patients, contrary to patients with type II CA. Generally, these patients have a stable neurological defect due to their “fixed” spinal cord dysplasia [34], and their motor deficit tends to parallel the extent of the bony abnormality. Conversely, sensory findings are much less well predictable from the radiographic appearance.

**Type II CA** (figure 24 and 25). If the whole or a part of the tail bud fails to develop but the true notochord is unaffected, primary neurulation occurs nor-

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**FIG. 22.** Caudal agenesis, type I: lumbar/thoracolumbar agenesis, 8-year-old girl. A. Radiograph, anteroposterior view shows agenesis of the lumbar and sacrococcygeal spine with ilioiliac approximation and reduced pelvic diameters. Only a rudiment of T12 articulating with an abnormal pair of ribs (arrowheads) is visible. B. Sagittal T1-weighted image shows rudiment of T12 articulating with an abnormal pair of ribs (arrowheads). The cauda equina also is visible. B. Sagittal T1-weighted image shows rudiment of T12 articulating with an abnormal pair of ribs (arrowheads).

**FIG. 23.** Caudal agenesis, type I. 8-year-old boy. A. Sagittal T1-weighted image and B. sagittal T2-weighted image show subtotal sacrococcygeal agenesis, with a rudiment of S1 as the last visible vertebra, articulating with medialized ileum (I). The cord terminus is blunt and lies opposite the lower half of L1 (arrow), a somewhat atypically “low” position for type I CA. There is terminal hydromyelia and “double bundle” arrangement of the nerve roots of the cauda equina. The dural sac tapers abruptly and ends abnormally high (arrowheads).

**FIG. 24.** Caudal agenesis, type I. 8-year-old girl. A. Sagittal T1-weighted image and B. sagittal T2-weighted image show subtotal sacrococcygeal agenesis, with a rudiment of S1 as the last visible vertebra, articulating with medialized ileum (I). The cord terminus is blunt and lies opposite the lower half of L1 (arrow), a somewhat atypically “low” position for type I CA. There is terminal hydromyelia and “double bundle” arrangement of the nerve roots of the cauda equina. The dural sac tapers abruptly and ends abnormally high (arrowheads).

**FIG. 25.** Caudal agenesis, type I. 8-year-old boy. A. Sagittal T1-weighted image and B. sagittal T2-weighted image show subtotal sacrococcygeal agenesis, with a rudiment of S1 as the last visible vertebra, articulating with medialized ileum (I). The cord terminus is blunt and lies opposite the lower half of L1 (arrow), a somewhat atypically “low” position for type I CA. There is terminal hydromyelia and “double bundle” arrangement of the nerve roots of the cauda equina. The dural sac tapers abruptly and ends abnormally high (arrowheads).
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mally, and there is interference only with the process of secondary neurulation [50]. There is a lesser degree of vertebral dysgenesis than in type I, with up to S4 present as the last vertebra. As a consequence, only the most caudal portion of the conus medullaris (corresponding to the metameres formed by secondary neurulation) is absent. In these cases, partial agenesis of the conus is difficult to recognize, because the conus itself is stretched caudally and tethered to a tight filum, lipoma [figure 24], terminal myelocystocele, or lipomyelomeningocele [figure 6]. In some cases, the cord tapers progressively to tether to the neck of an anterior sacral meningocele. In such cases, the anomaly may be discovered in later childhood or adolescence, when the patient develops constipation, urinary incontinence, dysmenorrhea, dyspareunia, or back pain. Teratomas or other caudal tumors can be found in a minority of patients. Low-back masses must be differentiated from overgrowing fatty tissue that is sometimes present at level of the buttocks in these patients. More often than in type I CA, imaging studies performed in these patients may be difficult to interpret, especially when looking for small teratomatous masses along the walls of anterior meningoceles, in the presacral space, or deep within the pelvic cavity. In these cases, presaturation slabs must not be placed anterior to the spinal column in order not to miss presacral malformed components.

Segmental spinal dysgenesis

The clinical-radiological definition of SSD includes (1) segmental agenesis or dysgenesis of the lumbar or thoracolumbar spine; (2) segmental abnormality of the underlying spinal cord and nerve roots; (3) congenital paraplegia or paraparesis; and (4) congenital lower limb deformities [48]. Segmental vertebral anomalies may involve the thoracolumbar, lumbar, or lumbosacral spine. As is the case with CA, the embryogenesis of SSD may be related to genetically induced notochordal derangement during gastrulation involving an intermediate, rather than the caudalmost, segment of the notochord [48].

In the most severe cases, the spinal cord at the level of the abnormality is thoroughly absent, and the bony spine is focally aplastic. As a result, the spine and spinal cord are “cut in two” (figure 25), with resulting acute angle kyphosis. Between the two spinal segments, the spinal canal is extremely narrowed or even totally interrupted. The lower spinal cord segment is invariably bulky and low-lying [48]. A horseshoe kidney is typically lodged in the concavity of the kyphosis [50]. Newborns with severe SSD are paraplegic at birth and invariably show hypotrophic and deformed lower limbs with equinocavovarus feet.

In less severe cases, there is focal hypoplasia of the spinal cord, which will therefore appear narrower than normal on MRI studies (figure 26) [48]. There is no disconnection of either the spinal cord or the spine, although bony stenosis of the spinal canal and minor vertebral abnormalities affect the pathological segment.

Fig. 24. – Caudal agenesis, type II. 3-month-old boy. Sagittal T1-weighted image shows the spinal cord is low and tethered (arrow) to an intradural lipoma (L). The vertebral anomaly is less severe than in type I, with S3 present in this case.

Fig. 24. – Agénésie caudale type II, garçon de 3 mois. Séquence pondérée en T2 – coupe sagittale. Moelle en position basse (flèche), lipome intradural (L). Les anomalies vertébrales sont moins sévères que dans le type I ; S3 est présent dans ce cas.

Fig. 25. – Segmental spinal dysgenesis, 2-month-old girl. Sagittal T2-weighted image shows acute thoracolumbar kyphosis with complete interruption of the spinal column. There are two completely separated spinal cord segments; the upper ends several vertebral levels above the gibbus (white arrowhead) and shows hydromyelia, whereas the lower is bulky and low (arrows). Notice extreme narrowing of spinal canal at the gibbus apex (black arrowheads).

Fig. 25. – Dysgénésie spinale segmentaire, fillette de 2 mois. Séquence pondérée en T2 – coupe sagittale. Cyphose thoraco-lombaire avec interruption complète du rachis. Il existe deux segments de moelle totalement séparés : la portion supérieure se termine à quelques niveaux vertébraux sous le gibbus (tête de flèche blanche et la partie inférieure est volumineuse (flèche). À noter le rétrécissement du canal rachidien faisant passer le gibbus par bosse à hauteur du sommet de la cyphose (flèche noire).
The MR picture in patients with spinal cord malformations may appear complicated and puzzling even to the experienced observer, we believe that a rational approach focusing on a correlation among clinical, embryological, and neuroradiological data greatly facilitates the diagnosis. Neuroradiologists should seek the maximum degree of collaboration with neurosurgeons and other specialists involved in the management of spina bifida patients in order to improve their diagnostic capabilities and to provide more useful information for the management of these children.

CONCLUSION

Although the MR picture in patients with spinal cord malformations may appear complicated and puzzling even to the experienced observer, we believe that a rational approach focusing on a correlation among clinical, embryological, and neuroradiological data greatly facilitates the diagnosis. Neuroradiologists should seek the maximum degree of collaboration with neurosurgeons and other specialists involved in the management of spina bifida patients in order to improve their diagnostic capabilities and to provide more useful information for the management of these children.

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