



Historical Perspective of Occult Spinal Dysraphism

1

Chad J. Jensen, Marc Vetter, Paul J. Choi, Rod J. Oskouian,
and R. Shane Tubbs

Introduction to Spinal Dysraphism

The first known account of spinal dysraphism was published during the seventeenth century. Anatomist Nicolaes Tulp (Fig. 1.1), famously portrayed in Rembrandt's painting "The Anatomy Lesson of Dr. Nicolaes Tulp," described congenital spinal abnormalities in his textbook *Observationes Medicae*. In the 1641 edition, he described six cases of spinal dysraphism and first introduced the term "spina bifida," literally "split spine." One of the cases, involving a lumbosacral lesion, was described as having "the prolongations of the nerves scattered in different directions through the tumor" [1]. In 1691, the Dutch surgeon Frederik Ruysch (Fig. 1.2) published ten case reports of spina bifida. Among other findings, Ruysch was the first to posit that a link existed between spina bifida and hydrocephalus [2]. Ruysch, like Tulp, recommended against surgical intervention because of the high morbidity and mortality, considering the condition inoperable [1]. There have been many subsequent publications on dysraphic states, but there are several disagreements about the pathogenesis and clinical importance of the condition and about

C. J. Jensen

Department of Anatomical Sciences, St. George's University, Grenada, West Indies

M. Vetter · R. S. Tubbs (✉)

Seattle Science Foundation, Seattle, WA, USA

e-mail: shamet@seattlesciencefoundation.org

P. J. Choi

Seattle Science Foundation, Seattle, WA, USA

National University of Ireland, Galway, Ireland

R. J. Oskouian

Swedish Neuroscience Institute, Seattle, WA, USA

© Springer Nature Switzerland AG 2019

R. S. Tubbs et al. (eds.), *Occult Spinal Dysraphism*,

https://doi.org/10.1007/978-3-030-10994-3_1

Fig. 1.2 Frederik Ruysch



French anatomist Jean Cruveilhier (Fig. 1.3) was the first physician to theorize, using anatomical and clinical evidence, that spinal dysraphism was the result of a developmental abnormality [2]. Under the category of spinal dysraphism fall a range of conditions varying in clinical and pathological severity. Regarding the definition and classification of dysraphic states, the first point is that the various macroscopic pathological findings distinguish specific subtypes of dysraphia. The scope of this chapter is *occult* spinal dysraphism (OSD), *occult* referring to that which is hidden. As noted by Tubbs et al. [4], these anomalies are almost entirely restricted to bony elements (vertebrae), and the underlying lesion is completely covered with the skin. This distinguishes OSD from its counterpart, *spina bifida cystica*. The occult dysraphic states include tethered cord syndrome, dilated spinal canal, posterior and anterior spina bifida, and split cord malformations [2]. Although the occult form remains covered with the skin, there can still be various external clues to an underlying pathology. Friedrich Daniel von Recklinghausen, a contemporary of Cruveilhier, was the first anatomist to document the association of lumbosacral hypertrichosis, one of these external clues, with spina bifida occulta [2]. Since then,

Fig. 1.3 Jean Cruveilhier

many other conspicuous anomalies have been clinically linked to occult spinal dysraphisms. Tubbs et al. describe the “enigmatic human tail,” noted in India and among peoples of remote islands in the Atlantic Ocean by Pliny and Pausanias, during the first and second centuries, respectively [4]. Although rare, such stories persisted through the centuries – their link to occult spinal dysraphisms still unknown. However, in a 2013 case report, Melo et al. [5] described three cases of human tails, all three of which showed underlying spinal dysraphism as revealed by magnetic resonance imaging. James and Lassman outlined other cutaneous findings such as subcutaneous lipomas, hypertrichosis as mentioned in the case by Virchow, angiomas, and dermal sinus tracts [6]. The growing documentation and understanding of conspicuous abnormalities associated with occult spinal dysraphisms has allowed medical professionals to more quickly render an effective diagnosis for patients with these conditions.

Spinal dysraphism was introduced into the medical lexicon by Friedrich Bremer, a German neurologist, in 1926 [7]. In 1940, Lichtenstein reintroduced the term to denote defective fusion of the neural tube in the dorsal midline during embryo development, which can cause phenotypic abnormalities in the following germ layers: somatic ectoderm (cutaneous), mesoderm, and neuroectoderm [8]. These present with various clinical manifestations involving abnormalities of the skin, muscles, vasculature, and nervous system. Lichtenstein drew attention to many cases that seemed more obscure and sometimes lacked the cutaneous or other external findings frequently seen with overt spina bifida: *occulta*, as it would come to be described. By definition, therefore, spinal dysraphism includes all forms of spina bifida: *occulta*, *aperta*, *anterior*, and *posterior* [9].

The designation *spina bifida occulta* is credited to Rudolf Virchow (Fig. 1.4). While the external lesions of meningoceles and myelomeningoceles are clearly visible, those of spina bifida *occulta* are not. In 1875 he addressed the *Berliner Gesellschaft für Anthropologie* regarding a case presented a few months earlier by Dr. Bernhard Ornstein in Athens. Dr. Ornstein had described a Greek soldier “whose loins presented an abundant crop of long hairs. The hairy growth occupied the middle line of the back, and extended thence on each side. The hairs were so long, that the man found it necessary periodically to cut them, in order to prevent interference with defecation” [10]. Virchow, who at the time was working in the

Fig. 1.4 Rudolf Virchow



Institute for Pathology at the Charité teaching hospital in Berlin, noted in his address that he had encountered a similar case of a 24-year-old female who had died from peritonitis secondary to typhoid fever. Virchow described a patch of hair measuring 10 cm in width, the hairs being 6–7 cm in length, with normal-appearing skin at the site. This patch, which he likened to the tuft of feathers on a Polish fowl and therefore called a “tuft of hair,” was associated with a palpable depression in the spine [10]. On dissection, Virchow found that the upper sacral vertebrae were “replaced by membrane” [10]. He opined at the time that the patch of hair was “the result of local irritation, due to disturbance during the development of the spinal column” [10].

While Virchow is credited with coining the term, cases had been documented during the seventeenth century (see above). Along with Tulp and Ruysch, Govert Bidloo (Fig. 1.5), an anatomist, playwright and student of Ruysch, published a case in 1708 in his surgical book *Exercitationum Anatomico-Chirurgicarum*, which was probably the first documented lipomyelomeningocele [11]. On autopsy, the conus

Fig. 1.5 Govert Bidloo



medullaris was found within the sacrum. Lipomyelomeningoceles are a form of OSD. According to Bosmia et al. [12], Johnson was credited by Rogers in 1971 with the first description of what was later called “lipomyelomeningocele.” However, it was in fact Bidloo, 172 years before Johnson, who described this form of dysraphism.

Soon after Virchow’s description, physicians began publishing case reports of spina bifida occulta and its associated clinical findings. Virchow’s own pupil, the aforementioned German pathologist Friedrich Daniel von Recklinghausen (Fig. 1.6), reported a case of a 9-year-old girl who had a tuft of hair in the lumbosacral region with a spinal depression and, on autopsy, confirmed the absence of the fifth lumbar spinal process. He described tethering of the cord at S2 by a fatty tumor and found that the fluid in spina bifida was indeed from the subarachnoid space [1]. The patient also suffered from chronic myelitis of the metatarsals in the left foot with an area of anesthesia. von Recklinghausen reported another case of a man with spina bifida occulta and club foot, a neurotic ulcer, and decreased sensation in the

Fig. 1.6 Friedrich von Recklinghausen



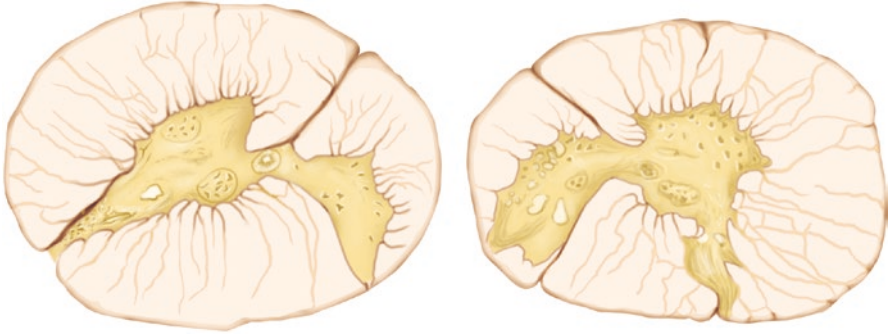


Fig. 1.7 Image of split cord malformation after von Recklinghausen

Fig. 1.8 Conrad Brunner



left foot (Fig. 1.7). Anatomist and physician Johann Conrad Brunner (Fig. 1.8), famous for his research on the pancreas and duodenum and identifying the eponymous “Brunner’s glands,” had presented a similar case years earlier, which also confirmed that spina bifida occulta is often associated with other physical findings

such as hypertrichosis and club foot [10]. In 1688, he published a case of a newborn with hydrocephalus and a cavity within it, noting a lumbosacral dysraphism and intramedullary cyst that was punctured by another physician, leading to the infant's death. Autopsy showed dysraphia as indicated by failure of fusion of the vertebral arches with myelomeningocele, hydrocephalus, and syringomyelia, described here 150 years before the term was coined. In his conclusion, Brunner believed that the syrinx was either a stand-alone malformation or part of the lumbosacral dysraphia [12].

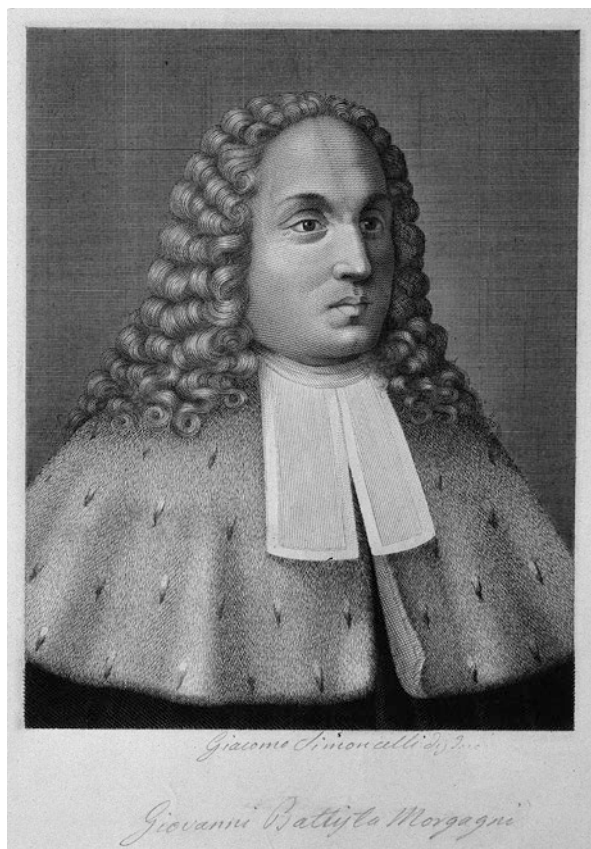
Giuseppe Muscatello (1866–1951), who had been a pupil of von Recklinghausen at the Institute of Pathology and is credited for his pioneering work in establishing cancer research as its own field and the first cancer center in Italy, also studied spina bifida aperta and occulta. In his various accounts of OSD, he found a dermoid cyst associated with the spinal lesion, other patients with the commonly seen focal tuft of hair, and some with split cords. He even stated his belief that some of these cases were hereditary [13]. These contributions helped establish spina bifida occulta as a clinical entity, sparking wide interest in its pathogenesis and, ultimately, its therapeutic options.

Forms of Occult Spinal Dysraphia

Tethered Cord and Filum Terminale Syndrome

Before Virchow used the term *spina bifida occulta*, a case in 1857, presented by Athol Johnson, a surgeon at the Hospital for Sick Children in Toronto, was probably the first documented instance of tethered cord syndrome, as it came to be known during the latter half of the twentieth century. The patient was a 10-month-old boy who experienced twitches and convulsive movements in the right leg. There was a swelling over the sacrum on which Johnson operated, discovering a lipoma emerging from the spinal canal and adhering to the membranes enveloping the spinal cord. Although documented surgical methods for the treatment of such a condition were at best anecdotal in 1857, Johnson successfully dissected the lipoma from the cord and the patient's twitches completely resolved after the operation [14]. Building upon the work of Johnson, in 1891, W.L. Jones performed what is now called the surgical "untethering" of the spinal cord in a 20-year-old male who had developed incontinence, club foot, and weakness and atrophy of the lower extremities. During his operation, Jones successfully dissected a dense fibrous band, stating that the "spine [was] trephined to relieve pressure on the cauda equine" [15]. The patient was able to walk without pain 6 months after the operation. Johnson and Jones' interventions began providing evidence that occult spinal dysraphisms could potentially be managed with surgery. This represents a break from previous medical opinions regarding the treatment of spinal dysraphism, which tended to advocate for nonintervention due to an extremely high rate of surgical complications. Renowned eighteenth century anatomist Giovanni Morgagni (Fig. 1.9) exemplified this line of thinking when presented with a case of spinal dysraphism in an infant, stating "The life of the little patient, however, is usually cut short by convulsions and other consequences of injury to the nerves; and these evils happen more speedily, if the nerves are pricked in opening the tumour" [2].

Fig. 1.9 Giovanni Morgagni



Not until 1940 did Lichtenstein first propose that tethering of the cord can cause paraplegia, though his hypothesis was not accepted at the time [16]. In 1953, Garceau (Fig. 1.10), an orthopedic surgeon, whose interest in congenital club foot and scoliosis, among other conditions, led him to discover what he termed *filum terminale syndrome* or *cord-traction syndrome*. Garceau had seen three cases, one with congenital scoliosis, one with idiopathic scoliosis, and one with tuberculosis of the spine, who all experienced lower extremity spastic paralysis. All three underwent laminectomy and resection of a thickened and tightened filum. In the patient with congenital scoliosis, the conus medullaris had remained in its fetal position, and in another patient the filum separated 1 cm after sectioning [17]. On the basis of his surgical findings and postoperative symptom improvement, Garceau posited that tension on the filum, exacerbated by spine flexion, for example, pulls on the hind-brain, displacing it downwards through the foramen magnum, essentially generating a Chiari malformation [15, 18, 19].

In 1976, Hoffman et al. [16] introduced the term *tethered spinal cord syndrome* in a publication on 31 patients who had motor and sensory deficits in the lower extremities. They confirmed that the symptoms improved after dividing a thickened filum terminale. Hoffman, along with Bruce Hendrick and Robin Humphreys, all

Fig. 1.10 George Garceau



three of whom worked at the Hospital for Sick Children in Toronto, advanced the ability of clinicians to recognize and diagnose *tethered spinal cord syndrome* by developing an anatomical definition for the condition. The “3 Hs,” as these physicians were collectively known, posited that a radiographic diagnosis of a filum terminale at least 2 mm in diameter and a low-positioned conus medullaris amounted to *tethered spinal cord syndrome* [2, 15, 16]. However, Yamada states that despite these advances, the establishment of *tethered spinal cord syndrome* was fraught with disagreement among physicians and scientists, partly because the evidence for it was based solely on visual findings at surgery [20].

Split Cord Malformation

In addition to tethered spinal cord as a type of OSD, the discovery of split cord malformation (SCM), formerly known as diastematomyelia, further extended the clinical entity. The earliest known case of SCM dates to circa 100 CE in Israel’s Negev Desert. The bones of a 20-year-old man were discovered in a tomb and found to have a butterfly vertebra with a bony process dividing the spinal canal into two halves at the thoracolumbar junction [21]. A number of cases were subsequently recorded, describing the same findings as “disjunction” of the cord, per Mathern, Peacock, and Perret in 1624. However, as noted by Saker et al. [21], it was not until 1837 that the pathologist Ollivier d’Angers (Fig. 1.11) coined the term *diastematomyelia*: Greek *diastema*, cleft, and *myelos*, spinal cord. von Recklinghausen, in addition to his aforementioned documentation of spina bifida occulta and associated

Fig. 1.11 Ollivier
d'Angers



clinical findings, remarked on SCM in a 31-year-old female with a split cord from the thoracolumbar spine to the cauda equina. In the ensuing years, Hertwig described the condition as two hemicords within their own dural sacs separated by a bony process – differentiating the finding from what he called *diplomylelia*, wherein the cord itself has been duplicated and there is no such sectioning by a bony process. Herren and Edwards disputed this distinction, stating on the basis of their findings from 42 autopsies that the cord is in fact duplicated [21]. The confusion among terms has persisted, highlighting the need for a more concise and accepted classification system.

In 1886, Humphry published findings from the dissections of six specimens with spina bifida, which account for early descriptions of diastematomyelia. One specimen was described as having deficiencies in the vertebral arches above the spina bifida with a notable projection of bony processes from the vertebral bodies, bisecting the spinal cord. In another specimen, Humphry found a similar presentation in lumbosacral spina bifida with a median process perforating the spinal cord directly above it and severe anatomical abnormality in the vertebral bodies of the lower spine [22]. In 1973, Gardner, whose work on dysraphic states was mentioned earlier, described diastematomyelia as an asymmetrical and longitudinal division of the spinal cord. He noted that at the spinal level involved there is often shortening, scoliosis, or fusion between vertebral bodies, the spinal canal is widened, and the

two hemicords are commonly contained within separate dural sacs and less commonly within the same one [2].

To avoid confusion of terminology and definitions, Pang et al. [23] set out to classify the condition succinctly, using the following three points as criteria for classification: the presence of two cords, the nature of the dural sacs, and the characteristics of the median septum. He distinguished type I and type II SCM, type I having two hemicords each within its own dural sac, separated by a midline bony septum, and type II having two hemicords within a single dural sac, separated by a nonrigid, fibrous median septum [23].

Dysraphia Since the Mid Twentieth Century

These dysraphic states were extensively researched during the 1940s by Ingraham, who documented associated congenital anomalies and provided criteria for surgical intervention. Through the 1950s to the 1970s, James and Lassman, inspired by the works of Ingraham, furthered understanding of OSD states in their authoritative book *Spinal Dysraphism: Spina Bifida Occulta* [9]. Their focus was spina bifida occulta, distinct from overt spina bifida or *aperta*. In a 1972 publication, they noted that spina bifida occulta had been “regarded as a curiosity rather than as an entity with possible clinical implications” as it was often discovered on routine X-rays but not remarked clinically [6].

James and Lassman relied on a distinction they attributed to Koch, whereby macroscopic surgical exploration of the spine revealed no herniation of neural contents [9]. During the preceding years, spinal surgical management had entailed high morbidity and mortality, particularly before the introduction of antibiotics in the 1940s. The authors noted an urgency to survey the literature and begin their own research to improve management and outcomes. James and Lassman suggest the following reasons for this urgency:

The clinical importance ... lies in the extrinsic anomalies which bind down the spinal cord or its nerve roots and prevent them from changing their position within the vertebral canal as they normally should to accommodate the growth of the vertebral column and of the spinal cord. If the spinal cord is tethered it will suffer a traction force during vertebral growth which it can accommodate to some degree in some cases by increasing its rate of growth, but when this compensatory reaction can do no more, the traction force will cause failure of neuronal conduction and ischemia owing to failure of blood supply or to venous congestion with possible thrombosis... It is all these factors which produce the changes in the lower limbs, bladder and bowel. [9]

Their research has since helped to establish recommendations for when surgical intervention is appropriate. It also initiated a thorough analysis of associated clinical findings in patients with spina bifida occulta, intended to help the physician in early diagnosis and consequently to achieve better clinical outcomes [9].

James and Lassman are particularly known for their work on tethered spinal cord syndrome, and in 1972 they published their findings and coined the term *meningocele manqué* (MM). Manqué (French) refers to that which is lacking; the authors posited that the bands are remnants of meningoceles that had failed to mature during embryo

development [24]. Various cases since James and Lassman's publication have challenged the claim that tethering bands terminate at the inner aspect of the dura. Although their definition did not provide a strict guideline for termination locations as a qualification for MM, they modified the definition in 1977, stating that bands can terminate outside the dura in defective laminae of the vertebrae or in the skin [24]. Rajpal et al. [25] also challenged the classification in 2007, setting out to establish a system that classed tethering tracts as "short tethering tracts" and "long tethering tracts," further distinguishing the two on the basis of histological features as having either epithelial or non-epithelial components. This eventually led to placing what would once have been classified under MM into distinct categories, i.e., dermal sinus tracts and limited dorsal myeloschisis. As observed by Schmidt et al. [24] there can be further subdivisions, but the authors note that MM is an entity in itself, adequately defined as "tethering of the spinal cord, nerve roots, and/or filum terminale by single and/or multiple aberrant nerve roots, fibrous bands, and/or adhesions, which terminate onto the dorsal dura mater, epidural space, or overlying lamina" [24].

Lastly, Dr. Shokei Yamada (1926–2017) (Fig. 1.12) who was a Professor and Chairman Emeritus for the Department of Neurosurgery, Loma Linda University School of Medicine, contributed significantly to our current understanding of the tethered cord. He developed an experimental model of tethered spinal cord and was editor for books on this topic [20].

Fig. 1.12 Shokei Yamada



Conclusion

Occult spinal dysraphism has an extensive history with a plethora of cases documented in the medical literature since the seventeenth century, and there are also cases from antiquity that we would class retrospectively as spinal dysraphism. Renowned pathologists such as Virchow and von Recklinghausen began an inquiry into cases of neurological deficits and apparent orthopedic cases such as club foot to initiate better understanding of this clinical entity, which Virchow termed spina bifida occulta. Furthermore, the work of more contemporary surgeons such as Athol Johnson, W.L. Jones, Harold Hoffman, and Shokei Yamada significantly advanced the ability of medical professionals to recognize, classify, and actively manage occult spinal dysraphisms. It is on the shoulders of these and other giants in the field that our current understanding of such neurological derailments is based.

References

1. Goodrich JT. A historical review of the surgical treatment of spina bifida. In: Ozek M, editor. *Spina bifida management and outcome*. Milan: Springer; 2008. p. 3–9.
2. Safavi-Abbasi S, Mapstone TB, Archer JB, Wilson C, Theodore N, Spetzler RF, Preul MC. History of the current understanding and management of tethered spinal cord. *J Neurosurg Spine*. 2016;25:78–87.
3. Gardner WJ. *The dysraphic states*. Amsterdam: ExcerptaMedica; 1973.
4. Tubbs RS, Malefant J, Loukas M, Oakes WJ, Oskouian RJ, Fries FN. Enigmatic human tails: a review of their history, embryology, classification, and clinical manifestations. *Clin Anat*. 2016;29:430–8.
5. Melo JRT, Pacheco P, Wanderley LE. Unusual spinal dysraphic lesions. *Case Rep Pediatr*. 2013;2013:210301.
6. James CCM, Lassman LP. Spinal dysraphism: an orthopedic syndrome in children accompanying occult forms. *Arch Dis Child*. 1959;35:315–27.
7. Pryse-Phillips W. *Companion to clinical neurology*. 3rd ed. New York: Oxford University Press; 2009. p. 134–311.
8. Lichtenstein BW. Spinal dysraphism: spina bifida and myelodysplasia. *Arch Neurol Psychiatr*. 1940;44(4):792–810.
9. James CC, Lassman LP. *Spinal dysraphism: spina bifida occulta*. London: Butterworths; 1972. p. 3–6.
10. Sutton JB. Abstract of a clinical lecture on spina bifida occulta, and its relation to ulcus perforans and pes varus. *Lancet*. 1887;2:4–7.
11. Tubbs RS, Cezayirli P, Blackerby WT. Govert Bidloo (1649–1713) and the first description of lipomyelomeningocele. *Childs Nerv Syst*. 2013;29:1219.
12. Bosmia AN, Tubbs RI, Clapp DC, et al. Johann Conrad Brunner (1653–1727) and the first description of syringomyelia. *Childs Nerv Syst*. 2014;30:193.
13. Tardieu GG, Loukas M, Fisahn C, et al. The Italian Giuseppe Muscatello (1866–1951) and his contributions to our understanding of childhood spina bifida aperta and occulta. *Childs Nerv Syst*. 2017;33:389.
14. Johnson A. Fatty tumour from the sacrum of a child, connected with the spinal membranes. *Trans Pathological Soc Lond*. 1857;8:16–8.
15. Bui CJ, Tubbs RS, Oakes WJ. Tethered cord syndrome in children: a review. *Neurosurg Focus*. 2007;23:E2.

16. Hoffman JF, Henrick EB, Humphreys RP. The tethered spinal cord: its protean manifestations, diagnosis and surgical correction. *Childs Brain*. 1976;2:145–55.
17. Saker E, Cox M, Loukas M, et al. George J. Garceau (1896–1977) and the first introduction of the “filum terminale syndrome”. *Childs Nerv Syst*. 2016;33(8):1233–6.
18. Garceau GJ. The filum terminale syndrome (the cord-traction syndrome). *J Bone Joint Surg*. 1953;35:711–6.
19. Lew SM, Kothbauer KF. Tethered cord syndrome: an updated review. *Pediatr Neurosurg*. 2007;43(3):236–48.
20. Yamada S. Introduction to tethered cord syndrome. In: *Tethered cord syndrome in children and adults*. 2nd ed. New York: Thieme Medical Publishers; 2010.
21. Saker E, Loukas M, Fisahn C, Oskouian RJ, Tubbs RS. Historical perspective of split cord malformations: a tale of two cords. *Pediatr Neurosurg*. 2017;52(1):1–5.
22. Humphry GM. Six specimens of spina bifida with bony projections from the bodies of the vertebrae into the vertebral canal. *J Anat Physiol*. 1886;20(Pt 4):584.1–592.
23. Pang D, Dias MS, Ahab-Barmada M. Split cord malformation: part I: a unified theory of embryogenesis for double spinal cord malformations. *Neurosurgery*. 1992;31(3):451–80.
24. Schmidt C, Bryant E, Iwanaga J, et al. Meningocele manqué: a comprehensive review of this enigmatic finding in occult spinal dysraphism. *Childs Nerv Syst*. 2017;33(7):1065–71.
25. Rajpal S, Salamat MS, Tubbs RS, Kelly DR, Oakes WJ, Iskandar BJ. Tethering tracts in spina bifida occulta: revisiting an established nomenclature. *J Neurosurg Spine*. 2007;7:315–22.