

# Pediatric Spinal Cord Diseases

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## PRACTICE GAPS

Spinal cord diseases in pediatric patients are highly variable in terms of presentation, pathology, and prognosis. Given that most of these diseases are treatable and typically nonlethal, early detection gives pediatric patients the best chance at fulfilling a normal, functional adult life long-term. Failure to detect these diseases until later may predispose patients to an adulthood with poorer neurologic function. Understanding the most common pediatric spinal cord diseases is, therefore, crucial in pediatric practice as it will assist clinicians in elucidating the niche differences in presentation and optimal management strategies.

## OBJECTIVES *After completing this article, readers should be able to:*

1. Recognize the most common presentation patterns of spinal cord diseases in pediatric patients.
2. Broadly describe the most common pediatric spinal cord diseases in terms of autoimmune, congenital, tumor, and vascular diseases.
3. Understand differences in epidemiology and imaging of different spinal cord diseases.
4. Engage patients and families about optimal treatment options and their relevant prognoses for different spinal cord diseases.

## ABSTRACT

Spinal cord diseases in pediatric patients are highly variable in terms of presentation, pathology, and prognosis. Not only do they differ with respect to each other but so too with their adult equivalents. Some of the most common diseases are autoimmune (ie, multiple sclerosis, acute disseminated encephalomyelitis, and acute transverse myelitis), congenital (ie, dysraphism with spina bifida, split cord malformation, and tethered cord syndrome), tumor (ie, juvenile pilocytic astrocytoma, ependymoma, and hemangioblastoma), and vascular (ie, cavernous malformations, arteriovenous malformations, and dural arteriovenous fistulas) in nature. These each require their own niche treatment paradigm and prognosis. Furthermore, presentation of different spinal cord diseases in children can be difficult to

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## ABBREVIATIONS

CNS central nervous system  
 CT computed tomography  
 MRI magnetic resonance imaging  
 MS multiple sclerosis  
 WHO World Health Organization

discern without epidemiologic and imaging data. Interpretation of these data is crucial to facilitating a timely and accurate diagnosis. Correspondingly, the aim of this review was to highlight the most pertinent features of the most common spinal cord diseases in the pediatric population.

## INTRODUCTION

There is a large burden of disease of the central nervous system (CNS) across the brain and spine, even when excluding headache, trauma, and back pain. (1) Spinal cord diseases alone are composed of an incredibly varied number of presentations and pathologies that can greatly impact survival and quality of life. When considering disease of the spinal cord, however, the pediatric population is distinct from the adult population for a variety of reasons. First, developmental abnormalities leading to spinal cord disease are more likely to manifest at a younger age. (2) Second, the neurobiology of a pediatric spinal cord renders it vulnerable to different insults that it can respond to in different ways. (3) Third, the prognosis of spinal cord disease must take into consideration a maturing spine in children, whereas in adults the spine has already matured. (4) In summary, understanding the broad array of pediatric spinal cord diseases must be done in tandem, but separate, to adult disease due to these distinctions. We herein summarize the most pertinent autoimmune, congenital, tumor, and vascular spinal cord diseases found in pediatric practice.

## INFLAMMATORY DISEASES

Inflammatory disease of the spinal cord can occur in response to multiple different stimuli, including infection, autoimmune disease, and syndromic sequelae. It is not uncommon for these spine diseases to co-present with intraxial cranial lesions. The most common diseases seen in children include multiple sclerosis (MS), acute disseminated encephalomyelitis, and acute transverse myelitis.

### Multiple Sclerosis

MS is an inflammatory disorder that manifests as white matter lesions spread over time and space in the CNS. There are many types of MS, with the most common being relapsing-remitting disease, which is overrepresented in pediatric cases compared with adult cases. Spinal cord involvement is almost universal in MS by the time of autopsy. (5)

**Epidemiology.** The global incidence of MS is approximately 33 per 100,000, with known geographic variance. In the higher-latitude countries of North America and Europe, the incidence can be as high as 140 per 100,000, and in the lower-latitude countries of Asia and Africa, as

low as 2 per 100,000. (6) Of all MS diagnoses, 3% to 10% first present during the adolescent years, with less during childhood. (7) Females are 2 to 3 times more likely to present with MS than are males.

**Presentation.** MS is characterized by episodic constellations of symptoms that can arise in a matter of hours to days. With respect to the spinal cord, extremity weakness and gait ataxia are the most common initial symptoms of MS. Spasticity in the lower extremities due to pyramidal tract involvement can also present. Involvement of the posterior column typically manifests as loss of proprioception. Other sensory deficits include paresthesia in the extremities and the Lhermitte sign, an electric shooting pain down the spine on neck flexion. There are other signs typical of MS outside the spine, including optic neuritis, leading to visual disturbances and vision loss, as well as mental disturbances and genitourinary symptoms.

**Imaging.** MS lesions are within white matter and are most commonly seen in the spine as focal plaques in the cervical region. These plaques are typically hyperintense and well-circumscribed on T2-weighted magnetic resonance imaging (MRI) and can be best made out on short tau inversion recovery sequencing. Unlike in the brain, hypointense lesions on T1-weighted MRI are not commonly seen in the spine.

**Treatment.** The goal of MS treatment is to maintain the functional status of the patient and reduce the risk of permanent neurologic disability. When there is an acute relapse, intravenous high-dose corticosteroids are the first-line intervention most commonly used. Other considerations in a refractory setting include adrenocorticotrophic hormone therapy and plasma exchange. (8) Longer-term, disease-modifying therapies such as interferon- $\beta$  and glatiramer have been used to provide long-term control. (5) Finally, adjunct symptomatic treatment can include medications, psychotherapy, and physical therapy to optimize activities of daily living. Surgery is rarely indicated in the management of MS.

**Prognosis.** The prognosis of MS has improved over decades. Today, many patients can expect to lead a largely normal life with optimized control for symptomatic episodes. More than 70% will initially present with the relapsing-remitting MS pattern that responds to therapy, although most of these patients will progress to secondary

progressive MS over time, losing independence and requiring more intensive management and care. (9)

### Acute Disseminated Encephalomyelitis

Acute disseminated encephalomyelitis is a diagnosis of exclusion. It describes an acute demyelinating process that is monophasic in nature, and it has been causally associated with previous viral infections. It has been proposed that an autoimmune reaction to myelin basic protein is the primary mechanism of this disease. (10)

**Epidemiology.** Acute disseminated encephalomyelitis is largely a childhood diagnosis, with the incidence estimated to be 0.1 to 0.3 per 100,000 children. (11)(12) Children are more affected than adolescents, and there is a male predominance with this disease. (13)

**Presentation.** Patients present with polyfocal neurologic symptoms, accompanied by encephalopathy that incorporates alterations in consciousness or behavior unexplainable by fever, systemic illness, or postictal course. (11) With respect to the spinal cord, symptoms include pyramidal signs, hemiparesis, and sensory loss. Other cranial sequelae are ataxia, optic neuritis, seizure, and speech impairment. The clinical course to maximal deficits is rapid and can occur within a matter of days. These symptoms then can fluctuate and evolve in severity over time, with 3 months typically implicated for diagnosis.

**Imaging.** Typically, patients present with radiographic evidence in the brain ranging from small punctate lesions to large tumefactive regions. In the spine, up to one-third of patients will demonstrate cord expansion as the most common feature of acute disseminated encephalomyelitis, and it typically occurs in the thoracic region. Across various sequences, MRI typically demonstrates heterogeneous, ill-defined lesions extending over a long segment of the cord (Fig 1). In pediatric patients, the presence of spinal lesions is often accompanied by the presence of cerebral lesions. (14)

**Treatment.** High-dose corticosteroids are the most widely accepted first-line treatment for acute disseminated encephalomyelitis. (15) Intravenous immunoglobulin treatment and plasma exchange are second-line treatments that have also been used in the case of refractory and fulminant disease. There is very rarely an indication for surgical intervention, although in cases of cerebral edema and cord compression, decompression may be required.

**Prognosis.** The prognosis of acute disseminated encephalomyelitis is largely favorable, with most patients making a full recovery. Typically, improvement in neurologic symptoms occurs within days of starting corticosteroid treatment, and they return to baseline within weeks rather



**Figure 1.** Sagittal proton density magnetic resonance image demonstrating characteristic confluent ill-defined intramedullary lesions in the spinal cord of a patient with acute disseminated encephalomyelitis.

than months. (11) Instances of long-term cognitive deficits have been reported in very young children, requiring life-long support. (16)

### Acute Transverse Myelitis

Acute transverse myelitis describes a clinical picture where there is acute, monophasic presentation of motor, sensory, and sphincter deficits that can be localized to the spinal cord bilaterally. (17) Etiologies are highly variable and can include viral myelitis, MS, acute disseminated encephalomyelitis, collagen vascular disease and paraneoplastic syndromes. However, in many cases, the cause is idiopathic. (18)

**Epidemiology.** The incidence of acute transverse myelitis in children has been estimated to be 1 to 2 per million children, and it is more than 4 times more common in adolescence as in childhood. (19)(20) Although males are more likely to present with acute transverse myelitis in general, females are more commonly affected in regions with known higher MS risk.

**Presentation.** The classic clinical presentation of acute transverse myelitis involves most commonly radicular and

back pain, followed by muscle weakness, sensory deficits (including paresthesia), and sphincter dysfunction. (21) These signs can at times be preceded by nonspecific fever and rash. The progression of these neurologic symptoms can be quite rapid, with maximal symptoms arising as early as 24 hours after the first symptom, but can climax as late as weeks after. One specific clinical presentation associated with acute transverse myelitis is neuromyelitis optica, a clinical triad of acute-onset optic neuritis, transverse myelitis, and positive anti-AQP4 antibody. (22) Spinal cord involvement typically extends beyond 3 vertebral levels in the cord, with cord swelling commonly seen. (23)

**Imaging.** The radiologic appearance of acute transverse myelitis is highly variable, with a centrally located hyperintensity causing cord enlargement, the most common imaging feature. There are times of extensive signal in the cord over multiple vertebral segments on T2-weighted MRI, with variable enhancement (Fig 2). Note that up to half of all patients with clinical acute transverse myelitis do not show any imaging abnormalities. (24)

**Treatment.** There is no standard of care for acute transverse myelitis. Acute administration of corticosteroids can provide relief in particular circumstances but is by no means universal. Treatment of known underlying etiology can sometimes provide symptomatic resolution. Plasma exchange and other forms of immune suppression can be considered if refractory to corticosteroid treatment. Finally, surgical decompression in cases of focal spinal cord enlargement is a treatment consideration should symptoms progress.

**Prognosis.** The prognosis after acute transverse myelitis is variable and seems to correlate with severity of initial symptoms. Large series that include pediatric patients indicate that fair to good functional recovery is likely in at least half, but not all, patients. (20)(25) Recovery occurred 4 weeks to 3 months after onset of initial symptoms, with no further improvement noted beyond 3 months. (20)

## CONGENITAL DISEASES

Congenital disorders of the spine are common in children. This is due to the complex process of neural tube and spinal cord development, which can be altered at many stages. As a result, a variety of congenital disorders can be detected in utero and in early infancy. Common disorders include dysraphism (spina bifida), split cord malformation, and tethered cord syndrome.

### Dysraphism

Dysraphism is a broad term that encompasses many spinal cord abnormalities resulting from failure of normal



**Figure 2.** Sagittal T2-weighted magnetic resonance image showing a long segment of abnormal hyperintensity and expansion of the visualized spinal cord, characteristic of transverse myelitis.

cord tissue differentiation, fusion, and closure. These defects typically present by birth, and they can involve unfused neural tissue (placode) in the spinal canal space (closed defect) or outside the spinal canal space (open defect). Of the most common presentations to a pediatric practice, myelomeningocele is an example of open spinal dysraphism, and meningocele is an example of closed spinal dysraphism. It should be noted that although the term *spina bifida* is often used interchangeably with *spinal dysraphism* in clinical practice, they are two distinct descriptions. In the strictest sense, spina bifida describes failed fusion of vertebral posterior elements and is often present with dysraphism. This consequent defect in the bony wall of the spinal canal allows neural tissue and meninges as



dysraphisms to physically extrude outside the spinal canal space. (26)

**Epidemiology.** The incidence of meningoceles and myelomeningoceles is approximately 1 to 5 per 1,000 live births (0.1%–0.5%) worldwide. (27)(28) This incidence is increased when the mother has insufficient prenatal folic acid levels. There may be a female predilection for these dysraphisms.

**Presentation.** On physical examination there can be a subcutaneous mass or lesion appreciated at the level of defect depending on whether the overlying skin is intact. In the case of myelomeningoceles, spinal cord neural tissue is displaced outside the spinal canal space (Fig 3). As a result, at birth, infants present with a wide array of intracranial symptoms due to spinal cord injury and hindbrain herniation. Acutely, patients can present with signs of brainstem compression, and later on, progressive lower limb paralysis and sensory loss, worsening gait, bladder and bowel dysfunction, and cognitive impairment. Hydrocephalus is very common in the first 6 months after birth.



**Figure 3.** Characteristic presentation of myelomeningocele in an infant with an open defect in the lumbar region.

(29) These signs and symptoms are less common in the case of meningoceles alone, as the neural tissue remains in the spinal canal space.

**Imaging.** Imaging modalities include ultrasonography and MRI. In the case of myelomeningoceles, imaging demonstrates a cerebrospinal fluid sac outside the spinal canal space, in which the spinal cord and meninges are displaced into posteriorly. In meningoceles, imaging will demonstrate similar findings except the spinal cord is not displaced and rather will continue its course in the spinal canal.

**Treatment.** The optimal timing to treat is not clear, particularly when a defect is detected before birth. Intrauterine closure prenatally is a rising trend, with it shown to decrease rates of hindbrain herniation and the need for ventricular shunting later on. (30) Yet there is an increased risk of preterm delivery, closure dehiscence, and abscess formation after this prenatal treatment. After birth, surgical closure of the defect remains the standard of care, with ventricular shunting a common adjunct.

**Prognosis.** The prognosis of spinal dysraphism without any treatment is poor, particularly in the case of myelomeningocele, where less than one-third of patients are expected to survive. With treatment, most patients survive to adulthood and will have a normal IQ. For myelomeningoceles, at least half of the patients are expected to be ambulatory but may require bracing to be so or require wheelchair support. Urinary incontinence is also a known sequela in a few patients.

### Tethered Cord Syndrome

Tethered cord syndrome describes a clinical picture resulting from an abnormally low-lying conus medullaris, typically associated with a short, thickened filum terminale, rendering the spinal cord immobile. This syndrome is seen in the setting of multiple neurologic, musculoskeletal, urologic, and gastrointestinal abnormalities, with myelomeningocele and lipomas being some of the most common. (31)

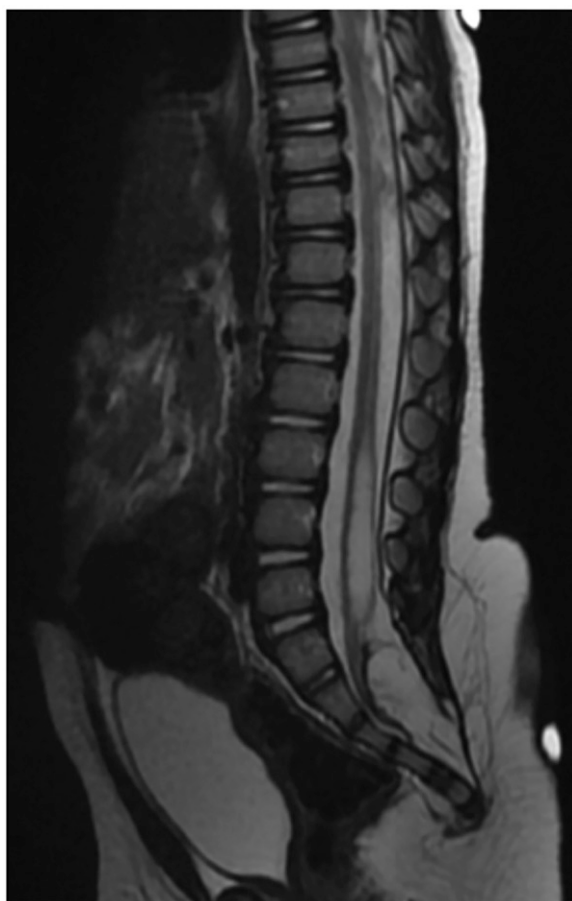
**Epidemiology.** The incidence of tethered cord syndrome is not well defined owing to its large number of etiologies and increase in incidental discovery after more accessible radiographic surveillance. (32) Conservatively, the incidence would be higher than the incidence of thickened filum terminale alone, which is approximately 12 per 100,000 children, and it is more commonly diagnosed in females than in males. (33)

**Presentation.** Tethered cord syndrome typically presents during childhood. Signs and symptoms include

neurocutaneous stigmata, bladder and bowel dysfunction, lower limb motor and sensory deficits leading to gait difficulty, lower limb length discrepancy, spinal deformity, and nonfocal lower limb pain. (34)

**Imaging.** In neonates and infants, ultrasonography can show abnormally thickened filum, a low-lying conus medullaris (below L2 after the first 3 months after birth), (35) and absent or diminished spinal cord movement. In older children, and after equivocal ultrasonography, MRI can be used to delineate fatty or fibrous infiltrate of the filum terminale, corresponding to hyperintense lesions on T1-weighted MRI and hypointense lesions on T2-weighted MRI (Fig 4).

**Treatment.** The standard of care for tethered cord syndrome is surgical untethering of the spinal cord by means of sectioning the filum via laminectomy. If there is a lipoma present, then excision of the mass is also indicated. Earlier treatment, in particular when other



**Figure 4.** Sagittal T2-weighted magnetic resonance image showing a low-lying tethered cord with a syrinx low-lying conus and a fatty filum terminale due to the presence of a lipoma.

progressive spinal comorbidities are present, portends to a more favorable functional outcome.

**Prognosis.** The prognosis after untethering is largely favorable in patients with tethered cord syndrome. Neurologic function is commonly stabilized or improved, and there is often resolution of pain. It is usually impossible to completely untether a cord, and so retethering may occur, requiring repeated untethering procedures. The issue of retethering diminishes once a child reaches late adolescence with a fully matured spine.

### Split Cord Malformation

Split cord malformation refers to a spectrum of spinal cord duplication anomalies. Two of the most common entities are diplomyelia and diastematomyelia. In the former there is complete duplication of the spinal cord into 2 hemicords, each with its own dural tube across the entire length of the spinal canal, whereas in the latter there is a region of division from 1 spinal cord into 2 hemicords in the same dural tube. (36) The hemicords can travel then in the same or a separate dural tube. Each hemicord contains its own central canal and can give off its own ventral and dorsal nerve roots. It is possible to have a bony septum between hemicords.

**Epidemiology.** The incidence of split cord malformation is poorly defined due to its extreme rarity, with cases in the literature limited to small case reports and series. It has been suggested based on association that it is less than 1 per 100,000. (37) Split cord malformation is more common in female patients than in male patients. (38)

**Presentation.** Although split cord malformation may be present from birth, patients often start to present during childhood. The first sign can be neurocutaneous stigmata, such as a hairy patch associated with concurrent spina bifida. (37) Other symptoms include back pain, motor and sensory deficits in the lower limbs, and skeletal deformity in the form of foot deformity and scoliosis. Bowel and bladder symptoms are common as well.

**Imaging.** Computed tomography (CT) can show widening of the interpedicular and spinal canal spaces and is particularly useful to visualize the osseous landscape and any possible interhemicord septum. MRI is able to distinguish between the 2 hemicords, in either a single or dual dural tube.

**Treatment.** Split cord malformation typically tethers the spinal cord and produces a tethered cord syndrome picture. As such, by the time of symptomatic presentation, surgical intervention is usually indicated based on expected disease progression. (39) In addition to untethering, surgery

involves removal of any intervening bony septa as well as the reconstitution of a single dural tube.

**Prognosis.** Symptoms typically stabilize and resolve after surgical treatment. Neurologic deterioration is unlikely but possible should retethering occur. It is possible for preexisting spinal deformities such as scoliosis to progress after split cord malformation has been treated, which require then their own treatment, often involving further surgery.

## TUMOR DISEASES

Intramedullary spinal cord tumors in pediatric patients are rare. Estimates suggest that up to 10% of all pediatric tumors in the CNS occur in the spinal cord, and the annual incidence is less than 1 case per 100,000 children. (40)(41) Per a large database study, the most common of these tumors is astrocytoma, specifically, juvenile pilocytic astrocytoma, followed by ependymoma and then heman-gioblastoma. (41)

### Juvenile Pilocytic Astrocytoma

Broadly, astrocytomas are tumors composed of astrocytic cells, and there exist many subtypes. The most common subtype in the setting of pediatric spinal cord tumors is juvenile pilocytic astrocytoma. These tumors are considered World Health Organization (WHO) grade I and portend to a better prognosis than the more infiltrating fibrillary or diffuse astrocytomas.

**Epidemiology.** Juvenile pilocytic astrocytoma is the most common CNS tumor in pediatric patients, with an incidence of 0.9 per 100,000. (42) Spinal juvenile pilocytic astrocytoma is as not common, however, with estimates suggesting that 2% of all juvenile pilocytic astrocytomas occur in the spinal cord. (43) The natural occurrence of juvenile pilocytic astrocytoma in the spine mirrors that of these tumors in general, with there being a slight male predilection and typical presentation during adolescence. (42)

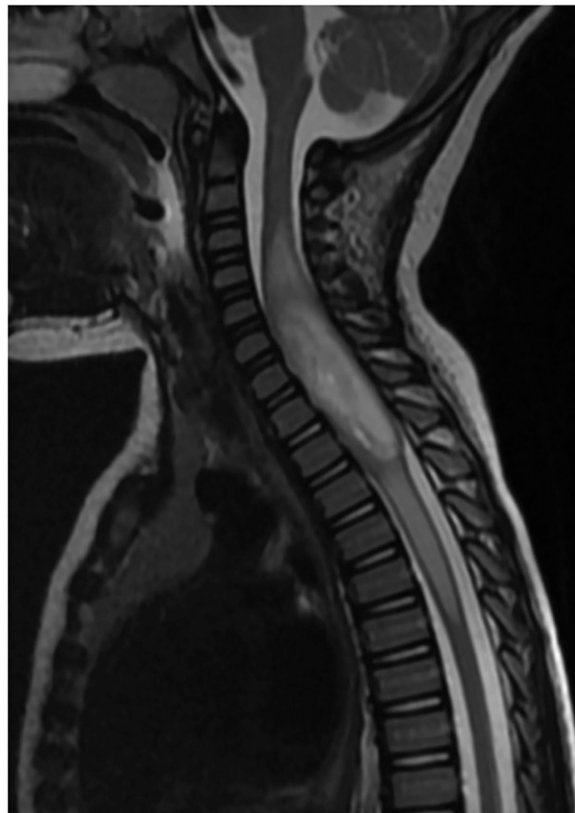
**Presentation.** Patients with juvenile pilocytic astrocytoma typically present months after symptom onset. (44) The most commonly reported locations of these lesions are in the cervical and thoracic spine. (45) The symptoms of these patients are consistent with intramedullary mass effect at their respective locations and include pain, motor and sensory deficits, falls, and sphincter dysfunction. (46) The degree of symptom severity at presentation typically correlates with the posttreatment outcomes.

**Imaging.** On CT there may be some evidence of spinal canal widening or pedicle erosion. On MRI, the expanded cord can demonstrate polar cystic components on T1-

weighted MRI; with variable intensity signal, the lesion is typically hyperintense on T2-weighted MRI (Fig 5). Although these tumors can appear radiographically similar to spinal ependymoma, juvenile pilocytic astrocytomas arise from the cord parenchyma and are more eccentrically located than ependymoma.

**Treatment.** The optimal treatment regimen for juvenile pilocytic astrocytoma involves maximal safe surgical resection. Although gross total resection is ideal, in contrast to spinal ependymomas, juvenile pilocytic astrocytoma often lacks a clear surgical plane, which is needed to facilitate clear gross total resection. (45) There is currently equipoise as to the benefit of adjuvant chemoradiotherapy after surgery given the low-grade nature of these lesions. Recent trends indicate that the use of chemotherapy continues to be part of standard care in these patients, with the use of radiotherapy becoming less popular with a less clear survival advantage in these patients. (45)(47)

**Prognosis.** The prognosis of spinal juvenile pilocytic astrocytoma is typically very good, and long-term survival is expected after surgical resection, with 1- and 5-year



**Figure 5.** Sagittal T2-weighted magnetic resonance image demonstrating a cervical juvenile pilocytic astrocytoma in an expanded spinal cord, with characteristic hyperintensity and polar cord signal change around the lesion.

survival rates estimated to be as high as 98% and 97%, respectively. (47)

### Ependymoma

Ependymal tumors are of neuroectodermal origin and sub-grouped based on histologic grade, location, and molecular profiling. Pertinent to the spinal cord are the 3 subgroups spinal subependymoma, WHO grade I; spinal myxopapillary ependymoma, WHO grade I; and spinal anaplastic ependymoma, WHO grade II/III; no group is more common in the pediatric spine than the others. (48)

**Epidemiology.** In terms of pediatric patients, ependymal tumors are proportionally more common than in adults, accounting for 5% of all primary CNS tumors at a rate of 0.3 per 100,000. (42) They are the most common spine tumors seen in children. (42) Among these tumors, less than half are found in the spinal cord, with there being a greater incidence in the adolescent age group compared with the children age group. (49) There is a slight predilection toward male patients.

**Presentation.** Patients can present up to 1 year after symptom onset. (50) Symptoms are often nonspecific and can vary based on a central cord syndrome, including pain and sensory disturbances. Lesions most commonly occur in the conus in children, and as such, pain in the lower extremities is the most common complaint. Sphincter dysfunction is common as well given the lower spine location typical of these lesions in children.

**Imaging.** There may be some evidence of spinal canal widening or pedicle erosion on CT. On MRI, the lesion is expected to be isodense to the cord on T1-weighted MRI, with well-delineated homogenous enhancement. This is clearer than the variable signal intensity associated with spinal astrocytomas, although both are expected to show hyperintensity on T2-weighted MRI (Fig 6). Given that these tumors arise from the ependyma lining the central canal, they are typically located in the center of the cord.

**Treatment.** The optimal treatment for spinal ependymoma is maximal safe resection, with recent surgical advances allowing for en bloc gross total resection over piecemeal subtotal resection. The strength of evidence in favor of adjuvant chemoradiotherapy remains low. In the case when gross total resection is not feasible due to anatomical limitations, radiotherapy may be used to extend progression-free survival at the least, although the optimal dose remains unclear. (51) For chemotherapy, small studies (52)(53) have suggested that chemotherapy may be safe



**Figure 6.** Sagittal T2-weighted magnetic resonance image of the cervical cord showing an ill-defined hyperintense intramedullary lesion at C5-6 characteristic of an ependymoma.

and effective to manage recurrence, although more robust studies are needed to validate this.

**Prognosis.** The prognosis of spinal ependymoma status post maximal surgical resection is excellent, with survival estimates as high as 100% at 5 and 10 years after diagnosis. (48) With respect to function, best outcomes are predicted by modest initial deficits with a less than 2-year history and gross total resection. (50)

### Hemangioblastoma

These tumors are low-grade, histologically slow-growing vascular lesions with neoplastic stromal cells and multiple capillary channels lined by a single layer of epithelium. Up to 80% of these lesions in the spinal cord will be associated with von Hippel–Lindau disease, and evaluation for such should also be included when hemangioblastoma is suspected. (54)

**Epidemiology.** Hemangioblastomas are very rare and constitute less than 1% of all CNS tumors in children at a rate of less than 0.1 per 100,000. (42) In these pediatric patients it is expected that up to 33% of these cases will occur in the spine if von Hippel–Lindau disease is present, and up to 10% when sporadic. (55) There seems to be no sex bias.



**Presentation.** Patients typically present with central cord syndrome signs based on the location of the lesion, with pain and weakness the most common symptoms over several months. (56) The most common spinal location for these tumors is the cervical spine. (57) These lesions may also be asymptomatic in the setting of von Hippel–Lindau disease that presents with other syndromic complaints.

**Imaging.** Similar to previous tumors, there may be some evidence of spinal canal widening or pedicle erosion on CT. On MRI, however, there will be a well-delineated cystic mass that is either hypointense or isointense with the spinal cord on T1-weighted MRI with an enhancing mural nodule and hyperintense on T2-weighted MRI. There may be associated syringomyelia as well.

**Treatment.** In the case of sporadic hemangioblastoma, gross total resection may be curative of the lesion. (57) However, due to the vascularity of this lesion, incision or core approaches cannot be pursued. Rather, a microsurgical technique similar to that for arteriovenous malformations should be used to maximize the extent of resection, with consideration for preoperative embolization and intraoperative hypotension as adjuncts. There is no role for adjuvant chemoradiation in the treatment of hemangioblastomas.

**Prognosis.** The prognosis after surgery for hemangioblastomas is very good. Complete resection reduces the risk of recurrence greatly, and normal overall survival is expected. Function-wise, patients are not expected to deteriorate from their preoperative baseline. (57)

## VASCULAR DISEASES

Vascular pathology in the spine represents a heterogeneous group of blood vessel disorders that arise and present as growing space-occupying lesions with abnormal arterial and venous shunting. It is, therefore, imperative to identify the nature of these intraspinal lesions to obtain angiographic evaluation. The most common vascular pathologies are cavernous malformations, arteriovenous malformations, and dural arteriovenous fistulas.

### Cavernous Malformation

Also known as cavernous angiomas, cavernous malformations are vascular hamartomatous lesions that contain immature blood vessels with dilated, thin tubular venous structures that are prone to internal bleeding. (58) Unlike arteriovenous malformations, there is no intervening spinal cord parenchyma in these lesions. In addition, these lesions can arise either de novo or secondary to radiation exposure.

**Epidemiology.** Across all spinal cavernous malformations in children and adults with an incidence of approximately 4 per 100,000 population, pediatric cases constitute approximately 10% of these cases. (59)(60) Among all pediatric intraspinal lesions, cavernous malformations constitute 2% of presentations. There is a distinct male preponderance in children, which contrasts the equal presentation in males and females in the case of adult spinal cavernous malformations.

**Presentation.** These patients typically present with signs of intraspinal mass effect. Progressive pain and weakness are the most common symptoms, with sensory deficits also reported. The cervical spine is the most common location in children, versus the thoracic spine in adults. (61) It is more common for these lesions to present with spinal hemorrhage in children than in adults, which is followed by acute neurologic decline. (62)

**Imaging.** CT can show a usually well-circumscribed lesion. On MRI, cavernous malformations appear as intramedullary lesions that typically demonstrate heterogeneous signal on both T1- and T2-weighted MRI, colloquially described as “popcorn” in appearance (Fig 7). There is also a hypodense rim on T2-weighted MRI due to hemosiderin deposition. Angiography is usually not required for confirmation.

**Treatment.** The decision to proceed with treatment depends on the symptom history and whether the lesion has bled previously. Surgical treatment is the standard of care in children when a lesion is symptomatic or the patient is clinically deteriorating; it is typically not indicated when the lesion has not yet bled. The surgical goal is to obtain complete resection, and because these lesions are not typically bloody, piecemeal excision is possible. The role of radiosurgery in treating these lesions in children remains controversial and undefined. (63)

**Prognosis.** When a spinal cavernous malformation is completely excised, the risk of recurrence is greatly reduced. It has been reported that recurrence of symptoms can still present in the case of brainstem lesions, even if complete excision is achieved. (64) Whether this applies to the spine is yet to be known. Function is expected to normalize back to baseline, but not certain.

### Arteriovenous Malformation

Arteriovenous malformations are a collection (nidus) of dilated blood vessels in which arterial blood is shunted into venous drainage without passing through normal interposed capillary beds. In the spine, the nidus of an arteriovenous malformation can be found in the intramedullary, extramedullary, or perimedullary space, depending on its type.



**Figure 7.** Sagittal T2-weighted magnetic resonance image demonstrating a well-circumscribed cervical lesion with a “popcorn” appearance classic for spinal cavernous malformation.

**Epidemiology.** Of all primary intraspinal masses, arteriovenous malformations constitute approximately 4% of presentations, with 20% of these occurring in the pediatric population, diagnosed more commonly in adolescence. (65) Spinal arteriovenous malformations are extremely rare, with fewer than 400 cases per year reported in the academic literature. (66) There may be a slight bias toward male patients. Also note that although arteriovenous malformations are known sequelae in rare hereditary hemorrhagic telangiectasia, which occurs in 1 per 10,000 pediatric population, they predominantly occur in the brain, with less than 2% occurring in the spine. (67)(68)

**Presentation.** Patients typically present months to years after experiencing the first symptoms. They describe a progressive course of neurologic decline starting from back pain. There is often associated sensory loss and lower extremity weakness as well. (69) In the case of subarachnoid hemorrhage of

arteriovenous malformations, patients also can present with sudden-onset myelopathy and excruciating back pain, which is more common in younger patients versus adults.

**Imaging.** On MRI there is an enlarged cord with heterogeneous signal coming from the nidus on T1-weighted MRI and contrast enhancement. On T2-weighted MRI, these lesions are hyperintense with flow voids. Confirmation can be obtained by angiography.

**Treatment.** Intervention for these lesions represents the patient’s best chance at symptom alleviation. However, the optimal and definitive treatment of spinal arteriovenous malformation in pediatric patients is unclear given the complex vascularity that some of these lesions possess. The efficacy and safety of traditional open surgery techniques for arteriovenous malformation resection have been compared with the more recent endovascular embolization techniques by many studies with no clear superior option. (70) In addition, radiosurgery has also emerged as another possible approach. (71) It is likely that the optimal approach to these lesions will be patient-dependent, incorporating a variety of factors such as size, location, number of feeders, and experience of the clinician in treating these lesions.

**Prognosis.** It seems that satisfactory functional outcome can be obtained after spinal arteriovenous malformation treatment in pediatric patients, irrespective of modality used. (70)(71)(72) Although not well studied in the pediatric spine, recurrence is possible, and its incidence seems directly correlative with incomplete lesion obliteration. (72)

### Dural Arteriovenous Fistula

A spinal dural arteriovenous fistula is a specific type of spinal arteriovenous malformation. It is a lesion with a single or multiple feeding arteries from a radiculomedullary artery forming a direct arteriovenous fistula shunt at the dural root sleeve with an intervertebral or radiculomedullary vein. Cord involvement can be distant to the fistula.

**Epidemiology.** Approximately 10% of all CNS arteriovenous shunts are estimated to occur in the spine, although it is believed that a much smaller percentage of this is to be the case in children. (73) Among all spinal dural arteriovenous fistula presentations that occur in up to 1 per 100,000 population, it is estimated that 20% will occur in the pediatric age group, with a known male predilection. (74)(75)

**Presentation.** These lesions occur most commonly in the cervicothoracic spine. Patients often present with a history of radicular pain and progressive

myeloradiculopathy that can have been present for many months due to venous congestion. (74) In the case of acute bleeding, which is less common, but more common than in adults, pediatric patients can report acute neurologic deficits such as motor, sensory, and sphincter deficits. Auscultation over the lesion may reveal a bruit.

**Imaging.** There can be an enlarged cord with multiple enhancing pial vessels on T1-weighted MRI. On T2-weighted MRI, the cord is also enlarged, with flow voids in the pia of the surface of the cord. Confirmation can be obtained by angiography.

**Treatment.** The optimal treatment of spinal dural arteriovenous fistula depends on multiple patient-dependent factors, with there being feasible arguments for both surgical resection and endovascular embolization. (73) These spinal dural arteriovenous fistulas are more likely to be cured than spinal arteriovenous malformation because they lack a nidus, making endovascular approaches more appealing in children. (76) Single arterial supply dural arteriovenous fistulas with slow ascending venous drainage are likely the most amenable to embolization compared with those with multiple feeders with giant venous ectasia and rapid drainage, which would be better exposed and removed by open surgery.

**Prognosis.** Prognosis of spinal dural arteriovenous fistula is typically very favorable. Pending complete elimination of the dural arteriovenous fistula irrespective of modality, recurrence is unlikely. Permanent functional deficits are also unlikely.

## RARE CONDITIONS

There are a variety of much rarer spinal conditions that have been reported in children. We highlight spinal cord ischemia and acute flaccid myelitis briefly herein. In addition, further study into degenerative cord disease in children is underway to determine its pertinence to the pediatric demographic, including genetic and acquired conditions in the setting of broad CNS diseases. (77)

### Spinal Cord Ischemia

Spinal cord ischemia occurs when the blood supply to the spinal cord is compromised, resulting in an ischemic insult. Depending on the anatomy of the arteries compromised, the spinal cord can be infarcted anteriorly, posteriorly, centrally, or, indeed, transversely. The primary causes of this in children are cardiac abnormalities causing hypotension and minor trauma causing fibrocartilage emboli. (78)(79) There have been only 25 cases in pediatric patients reported to date. (80) Typically these patients will

present acutely, with maximum symptoms in the first 72 hours after onset. (81) The most common symptoms are severe back pain and incontinence before motor deficits and myelopathy. (82) Consistent with a stroke diagnosis, spinal cord ischemia is best seen as increased T2 signal in the spinal cord on MRI. Treatment is primarily symptomatic in nature, with the role of decompression surgery and corticosteroids undefined. Prognosis is highly variable, as return of the abilities to walk and remain continent is estimated to occur in approximately half of all patients. (82)

### Acute Flaccid Myelitis

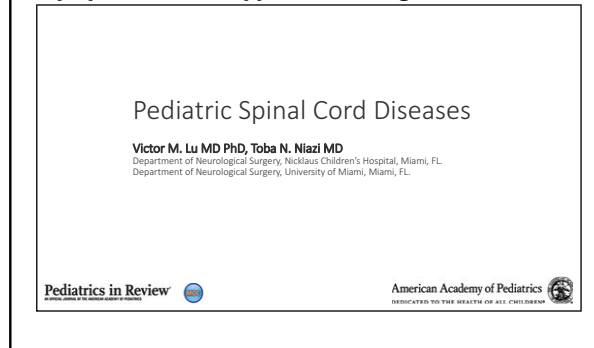
Acute flaccid myelitis is a mostly pediatric syndrome that describes a variable pattern of extremity weakness caused by a loss or dysfunction of anterior horn cells in the spinal cord. It is associated with infectious and inflammatory processes, the most well-known historically being poliomyelitis, and more contemporaneously enterovirus D68. (83) The incidence of acute flaccid myelitis seems to trend biennially and in clusters, with national estimates in the United States of less than 150 cases per year during the past decade. (84) Patients with acute flaccid myelitis typically describe upper respiratory symptoms or fever in the days preceding the onset of weakness, which once started can maximize neurologically over hours to days. (85) Weakness ranges from slight single-limb paresis to complete tetraplegia. (83) MRI typically shows gray matter–predominant, T2-hyperintense spinal cord signal abnormality, although this can be delayed up to 72 hours after weakness onset. (86) Treatment is largely supportive in nature via means of physical therapy, with anecdotal use of intravenous immunoglobulin, high-dose glucocorticoids, plasma change, and nerve transfer of lower motor neurons all described. (83)(87) The prognosis of this disease with respect to return of full function is poor, with estimates suggesting that only 1 in 4 patients will achieve this. (88) Most of these pediatric patients will proceed to experience multiple lifelong residual physical and psychosocial issues. (89)

## Summary

- Spinal cord diseases in the pediatric population are diverse in presentation and pathology.
- Autoimmune diseases tend to present throughout childhood, with management largely conservative in nature. Long-term follow-up is often needed.
- Congenital diseases manifest early in childhood, more in females than males, and often can be resolved with surgical treatment.

- Tumor diseases often present during adolescence, with a slight preponderance for male patients. Surgical resection and adjuvant therapy remain the standard of care, and prognosis varies depending on histology.
- Vascular diseases are rare and typically declare themselves during adolescence. Surgical treatment typically resolves these lesions.
- Given the unique niche age group, understanding of how common and rare pediatric spinal cord diseases manifest and can be managed will best equip professionals to provide the best care possible for the patient in both the short- and long-term.

**To view teaching slides that accompany this article, visit <http://pedsinreview.aappublications.org/content/42/9/486>.**



References for this article can be found at <http://pedsinreview.aappublications.org/content/42/No. 9/486>.





1. You are preparing to see a child in your clinic who was recently diagnosed as having a spinal cord lesion on magnetic resonance imaging (MRI). The family history includes an uncle who also had a “condition” of the spinal cord, although details are unknown to the child’s parents. As you consider how to counsel the family, which of the following is a key distinction between pediatric and adult spinal cord disease?
  - A. Developmental abnormalities causing spinal cord problems are more likely to manifest in adulthood than in childhood.
  - B. Pediatric and adult patients manifest identical responses to spinal cord problems.
  - C. Pediatric spinal cord biology makes it vulnerable to different injuries than adult biology.
  - D. Prognosis in children is determined by their already matured state of the spine.
  - E. Understanding of pediatric spinal cord diseases must be completely independent of that of adult diseases.
  
2. A 12-year-old girl well-known to your clinic presents for a follow-up after recent hospitalization. She had been admitted with bilateral leg weakness and trouble walking that worsened over several days. In retrospect, her family remembered that a similar but less severe episode occurred the previous summer. An MRI showed multiple lesions of varying ages in both the brain and the spinal cord. A diagnosis of multiple sclerosis was made. Which of the following types of multiple sclerosis is most likely to occur in a pediatric patient?
  - A. Infantile-onset disease.
  - B. Multiple sclerosis that never responds to therapy.
  - C. Primary progressive disease.
  - D. Relapsing-remitting disease.
  - E. Spinal cord–sparing disease.
  
3. You are seeing in your clinic an 11-year-old boy who has been previously healthy. His parents have brought him for evaluation because of worsening bladder dysfunction. This was initially noted a year earlier, and during the past months has worsened. The child has also complained intermittently of leg pain, with no clear inciting injury. Physical examination reveals bilateral sensory deficits in the lower extremities. You suspect tethered cord syndrome. Which of the following is associated with a favorable functional outcome?
  - A. Early surgical treatment.
  - B. In utero repair.
  - C. Preservation of associated lipoma.
  - D. Retethering of the cord.
  - E. Use of intravenous corticosteroids.

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4. A 9-year-old boy with a few months' history of lower extremity pain and weakness is admitted to the pediatric ward after computed tomography reveals a new spinal canal lesion with pedicle erosion. An MRI during the admission shows a well-delineated cystic mass in the spinal cord and features consistent with hemangioblastoma. Evaluation for which of the following conditions should be undertaken given suspicion for hemangioblastoma?
- A. Acute disseminated encephalomyelitis.
  - B. Brainstem herniation.
  - C. Diastematomyelia.
  - D. Hydrocephalus.
  - E. Von Hippel–Lindau disease.
5. During your urgent care shift you see a 7-year-old boy who is having difficulty using his left arm. His mother explains that he is a generally healthy child. He had a cold with low-grade fever the previous week but had already recovered before the day of presentation. The night before the visit the child had complained of trouble using his hand on the affected side; by this afternoon he was largely unable to use the entire arm. He does not complain of any pain or any other symptoms. He now has a flaccid paresis on examination with no-antigravity strength in the left upper extremity. The remainder of his examination findings are normal. Which of the following diagnoses is most likely to account for this child's presentation?
- A. Acute flaccid myelitis.
  - B. Juvenile pilocytic astrocytoma.
  - C. Myelomeningocele.
  - D. Split cord malformation.
  - E. Tethered cord syndrome.