Combined surgical and endovascular treatment of complex high-flow conus medullaris arteriovenous fistula associated with Parkes Weber syndrome: case report

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Parkes Weber syndrome (PWS) is a congenital overgrowth disorder characterized by unilateral limb and axial hypertrophy, capillary malformations of the skin, and high-flow arteriovenous fistulas (AVFs). Spinal AVFs in the setting of PWS are challenging vascular lesions that often contain multiple arteriovenous (AV) shunts. The present case report highlights an adolescent girl with PWS who presented with a ruptured complex high-flow conus medullaris AVF. She was successfully treated with endovascular embolization and microsurgery. At the 2-year follow-up, the patient remained free of neurological symptoms and had no recurrence of the vascular malformation.

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KEY WORDS Parkes Weber Syndrome; vascular malformation; arteriovenous fistula; treatment; spinal cord; endovascular; vascular disorders

PARKES Weber syndrome (PWS) is a rare congenital overgrowth disorder characterized by limb, axial, and visceral hypertrophy; capillary malformations of the skin; and the presence of significant arteriovenous (AV) shunting/high-flow vascular malformations.^{11,15,16} It is a heterogeneous disorder and most often arises sporadically, although familial cases have been described.² A genetic link related to a mutation in the *RASA1* gene has been reported to account for familial PWS. This variant is seen primarily in patients with multiple capillary malformations.¹⁰

Spinal vascular malformations in the setting of PWS are typically complex, exhibiting multiple AV shunts that demand a multidisciplinary approach because of the unique angioarchitectural characteristics (including the dural and pial feeders in the same lesion). Successful treatment hinges on surgical and/or endovascular interventions to disconnect all fistulous points. We report the case of an adolescent girl with PWS who presented with an acute, nonaneurysmal subarachnoid hemorrhage (SAH) due to a ruptured complex conus medullaris AV fistula (AVF) with multiple AV shunts. She was successfully treated using endovascular embolization and microsurgery.

Case Report

History and Physical Examination

A 14-year-old girl presented with abrupt-onset severe headache and vomiting, followed by a generalized seizure. CT scanning revealed diffuse SAH, predominantly in the posterior fossa, and mild hydrocephalus. Cerebral angiography demonstrated no intracranial aneurysm. Her prior surgical history included an epiphysiodesis procedure to help prevent progressive lengthening of her hypertrophied left leg. She had also undergone surgical removal of varicose veins and venous malformations in the same limb.

Physical examination revealed left lower-extremity soft-tissue and bony hypertrophy with multiple capillary malformations and hypertrophy of the left breast. Abdominal sonography and MRI demonstrated an asymptomatic left renal AV malformation (AVM). MRI of the spine showed a vascular malformation within the conus medullaris, which was suspicious for an AVM or an AVF, with associated acute hemorrhage within the cord terminus and SAH that extended to the lower lumbar thecal sac (Fig. 1). Spinal angiography revealed a prominent high-flow, intradural, ventral AVF fed primarily by the anterior

ABBREVIATIONS AV = arteriovenous; AVF = AV fistula; AVM = AV malformation; DSA = digital subtraction angiography; KTS = Klippel-Trenaunay syndrome; NBCA = *N*-butyl cyanoacrylate; PWS = Parkes Weber syndrome; SAH = subarachnoid hemorrhage.

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FIG. 1. Sagittal (A) and coronal (B) T2-weighted MR images of the lumbar spine, demonstrating multiple dilated perimedullary veins within the conus medullaris (yellow arrows). A serpiginous draining vein is also seen ascending from the L4–5 level to the conus medullaris (red arrows). Note the markedly enlarged left kidney with multiple dilated flow voids secondary to a renal AVM on the coronal image. Figure is available in color online only.

spinal artery (with a high-flow pial component) arising from the right T-11 intercostal artery (Fig. 2). A second intradural dorsal arterial feeder arising from the posterior spinal artery through the left L-1 lumbar artery was also identified (Fig. 3). The fistula point was located at the transition of the T-12 and L-1 vertebral levels (conus medullaris).

An additional intradural dorsal (slow-flow) AVF arising from the radicular branch of the left L-4 lumbar artery was identified with an ascending serpiginous drain-

ing vein up to the fistula point, at the level of the conus medullaris (Fig. 4). There were dilated varicose veins at the fistula point that resembled an intramedullary AV nidus (a pseudonidus angiographic appearance). The fistula finally drained into a long, descending, extradural vein, which then drained into the left common iliac vein. Findings were consistent with the diagnosis of an overgrowth disorder, with phenotypic features of PWS.

Treatment

During surgery we observed a recent hemorrhage around the spinal cord vascular malformation. The patient underwent bilateral T10–11 laminectomies and surgical ligation of a posterior fistula at the pseudonidus point. Postoperative spinal angiography showed residual filling of the vascular malformation. The patient then underwent endovascular embolization of the right T-11 and left L-1 arterial feeders using *N*-butyl cyanoacrylate (NBCA) and the anterior spinal artery was preserved. Repeat spinal angiography showed persistence of the L-4 intradural dorsal (slow-flow) component, which also required NBCA embolization; this achieved complete cure of the complex fistula (Fig. 5).

Outcome and Follow-Up

The patient tolerated the surgical and endovascular treatments well, without complications. After 2 years of follow-up, she remained free of neurological symptoms and was cured of the complex AVF.

Discussion

Our patient's syndrome and presentation are exceedingly rare—spontaneous, nonaneurysmal SAH secondary

FIG. 2. Anteroposterior (A) and lateral (B) DSA studies of the right T-11 intercostal artery, demonstrating the anterior spinal artery (yellow arrows) feeding the fistula point that has a pseudonidus appearance at the conus medullaris (red arrows) and drains into an arterialized extradural vein (white arrows). Corresponding lateral radiograph (C) with microcatheter within the anterior spinal artery, confirming the ventral location of this intradural fistula (green arrow). Figure is available in color online only.

FIG. 3. Anteroposterior (A) and lateral (B) DSA studies showing the intradural dorsal component of the conus medullaris fistula through the left posterior spinal artery (yellow arrows). Note reflux into the anterior spinal artery (red arrows). The anterior and posterior spinal arteries in this image “sandwich” the conus medullaris. Figure is available in color online only.

to a complex, high-flow conus medullaris AVF in the context of PWS. Spinal cord AVFs can be particularly challenging to treat in patients with PWS because the fistula point can have multiple feeders, and multiple fistulas can be present in a single patient.

Our review of the literature found no other cases in which authors describe the unique association of PWS with multiple intraspinal AVFs. There have been, however, 5 case reports in which this condition was characterized as Klippel-Trenaunay-Weber syndrome. Research supports the fact that Klippel-Trenaunay syndrome (KTS) and PWS are in fact 2 independent disorders, and therefore it is inaccurate to group these syndromes together as a single entity.

One case report described a 13-year-old girl who presented with myelopathy and 4 AVFs.⁷ The lesion was successfully treated with platinum fiber coils. Rohany and colleagues reported on a 37-year-old woman presenting with perineal and right lower-extremity radicular pain.¹¹ She was found to have 3 concurrent spinal perimedullary fistulas, which were treated with both embolization (of a large varix with platinum coil/NBCA mixture) and surgery (coagulation of multiple small AVFs and varix excision). Sharma published a case of a 16-year-old boy presenting with bilateral lower-extremity weakness and back pain.¹² This patient was found to have multifocal intradural spinal AVFs and bilateral renal artery aneurysms. The treatment was not described in this case. Tokunaga and colleagues presented a case of a 7-year-old girl with multiple perimedullary AVFs, which were treated successfully using a combination of transarterial and transvenous embolization.¹⁴ Finally, Iizuka and colleagues described the case of a 24-year-old woman with a conus medullaris spinal AVM who presented twice with SAH.⁴ She underwent transarterial glue embolization.

FIG. 4. Anteroposterior (A) and lateral (B) DSA studies of the left L-4 lumbar artery, demonstrating a radiculomedullary fistula with an ascending intradural dorsal serpiginous dilated vein (yellow arrows) up to the conus medullaris that further drains into the arterialized extradural vein (red arrows). Figure is available in color online only.

Differential Diagnostic Considerations

KTS and PWS are similar limb-overgrowth disorders characterized by complex vascular anomalies. These conditions have frequently been grouped together in the medical literature under the misnomer Klippel-Trenaunay-Weber syndrome, despite important differences. Since KTS and PWS are distinct entities, the grouping together of these conditions using the name Klippel-Trenaunay-Weber syndrome should be strongly discouraged.

Significant AV shunting/high-flow vascular malformations characterize PWS, whereas KTS is strictly a condition consisting of slow-flow malformations (including lymphatic, capillary, and venous vascular anomalies).^{1,18} Conflating the two syndromes fails to recognize vascular biological and genetic differences. Differentiating between KTS and PWS is essential, since they differ markedly in their clinical manifestations and unique treatment approaches.^{6,8}

Though PWS was initially thought to be a sporadic disorder, genetic links have been found to a mutation in *RASA1*. This may also be seen in other fast-flow vascular anomalies, including capillary malformation-AVMs and vein of Galen aneurysmal malformations. This mutation helps distinguish PWS from other overgrowth disorders such as hereditary hemorrhagic telangiectasia with high-flow AVFs, *PTEN* hamartomatous tumor syndrome, and KTS.^{6,9,10} This gene mutation similarly has been associated with spinal AV anomalies in patients with multifocal capillary malformations.¹³ Thus, genetic testing may be helpful to ensure the correct diagnosis in certain patients if it remains unclear clinically and can thereby help direct management and prompt further investigation for spinal vascular malformations. Genetic testing was not performed in our patient because of the inability by the parents to afford

FIG. 5. After superselective embolization with NBCA, DSA studies of the right T-11 intercostal artery (A), left L-1 lumbar artery (B), and left L-4 lumbar artery (C) showed complete obliteration of the fistulas without opacification of the pseudonidus centered at the conus medullaris.

the cost. However, we did not feel it was necessary as our diagnosis was straightforward.

Workup and Treatment Considerations

The yield of spinal axis imaging in nonaneurysmal SAH is low, particularly in perimesencephalic SAH.³ However, imaging of the spine should be considered in certain patients. At our institution, we perform MRI of the spine particularly when cerebral digital subtraction angiography (DSA) is nonrevealing (especially after 2 or more consecutive negative cerebral DSA studies) and in the following cases: posterior fossa–predominant SAH, certain predisposing vascular disorders, and pediatric patients because AVMs are more commonly a source of SAH when compared with the adult population.

When evaluating spinal vascular malformations in PWS, angiographic assessment should be thorough, as these lesions usually contain multiple fistulas. Endovascular angiography necessitates the evaluation of multiple visceral arteries to identify all potential “feeders” to the fistula. Careful assessment allows for precise classification of the lesions; this is essential to identify both the anterior and posterior spinal arteries and for treatment planning.

The classification of spinal AVMs and AVFs has evolved and has been under constant revision. The modified classification system proposed by Kim and Spetzler is based on anatomical and pathophysiological features of these lesions. Our patient had both intradural ventral and

dorsal fistulas with large dilated veins within the conus medullaris, compatible with the complex angioarchitecture of a conus medullaris AVM in their classification.⁵

If left untreated, these patients often experience progressive neurological decline or recurring acute hemorrhagic events, including spinal cord hematoma and SAH. Although there are no consensus guidelines for the treatment of intraspinal AVFs in patients with PWS, treatment should be individualized based on the angioarchitecture of the lesion and treatment goals set for the patient. Complete obliteration of the lesion may be challenging, and treatment should focus on clinical improvement and reducing the volume of the malformation.

Wilson and colleagues have demonstrated excellent long-term outcomes using combined endovascular and microsurgical treatments of conus medullaris AVMs, with 86% of patients remaining neurologically stable or improving over the long term.¹⁷ Similarly, a multimodality approach resulted in a positive outcome in our patient without complications.

The treatment for our patient was thoroughly discussed among the neurosurgery, neurointervention, and neurology services. It was deemed that the presence of multiple arterial feeders conveying to a pseudonidal point at the level of the conus medullaris was more amenable to a surgical ligation, particularly the posterior arterial feeders, rather than multiple embolizations through the arterial side. It was thought the surgical ligation would offer a more durable solution without the need of exposing the

young girl to repetitive endovascular treatments and the stochastic risks of excess radiation.

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Disclosures

The authors report no conflict of interest concerning the materials or methods used in this study or the findings specified in this paper.

Author Contributions

Conception and design: Bagherpour, Rodriguez, Moorthy, Maud. Drafting the article: Bagherpour, Maud. Critically revising the article: all authors. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: Bagherpour. Administrative/technical/material support: Bagherpour, Rodriguez, Maud. Study supervision: Maud.

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