Endovascular treatment of spinal arteriovenous fistula in a young child with hereditary hemorrhagic telangiectasia

Case report

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Hereditary hemorrhagic telangiectasia (HHT or Osler-Weber-Rendu syndrome) can manifest as sudden onset of epis-taxis or neurological deficit in a child with characteristic mucocutaneous telangiectasias or as an asymptomatic bruit with or without overlying cutaneous vascular lesions. The authors present a case study of a pediatric patient with HHT in whom a screening computerized tomography (CT) scan of the chest revealed an asymptomatic arteriovenous malformation (AVM) of the spine.

An 18-month-old child with a strong family history of HHT, including fatal central nervous system (CNS) hemorrhage and pulmonary AVMs, presented with a cutaneous telangiectasia of the pinna. The child was subsequently screened for potentially morbid pulmonary and CNS AVMs by using chest CT scanning and brain magnetic resonance (MR) imaging. A spinal MR image revealed a perimedullary macro-AVM (MAVF) resulting in a large venous varix within the parenchyma of the thoracic spinal cord. A transarterial embolization of the fistula was performed using N-butyl cyanoacrylate and ethiodol. Postembolization angiography confirmed obliteration of the fistula, and MR imaging revealed thrombosis and reduction in size of the venous varix. There were no neurological sequelae due to the treatment.

In families with HHT and a high risk of sudden severe morbidity or death from undisclosed pulmonary or CNS AVMs, screening chest CT scanning and CNS MR imaging should be considered. Interdisciplinary teams of neurosurgery and interventional radiology specialists should evaluate and treat such patients by using diagnostic and therapeutic angiography and, if necessary, surgery.

KEY WORDS • hereditary hemorrhagic telangiectasia • Osler-Weber-Rendu syndrome • endovascular embolization • pediatric neurosurgery

HEREDITARY hemorrhagic telangiectasia (also known as Osler-Weber-Rendu syndrome) is an autosomal dominant disorder of high penetrance (97%), with variable expressivity.2 Patients with HHT are characterized by the presence of multiple mucocutaneous telangiectasias and visceral AVMs.3,7 Type 1 HHT is caused by a mutation in the gene encoding endoglin, a receptor for a transforming growth factor–β (chromosome 9q34.1).1,15 Type 2 HHT is caused by a mutation of the activin receptor-like kinase 1, also a transforming growth factor–β receptor family member (chromosome 12q13).1,3,5,15 Both activin receptor-like kinase 1 and endoglin are expressed on endothelial cells and play a regulatory role in angio-genesis.

Common sites for the development of AVMs in HHT are the lungs, CNS, and liver. Although less common than cerebral AVMs in HHT, spinal cord AVMs can be the presenting lesion in children with this disorder. Spinal AVMs14 in patients with HHT are characteristically subpial perimedullary MAVFs.4,11 Macroarteriovenous fistulas can present as acute paralysis as a result of parenchymal or subarachnoid hemorrhage, painful acute myelopathy, or with insidious onset of sensorimotor neurological deficits or gait disturbance. In the absence of neurological symptoms, they can also be discovered through radiological screening, or the investigation of overlying cutaneous vascular lesions or bruits. Their therapeutic treatment in patients with HHT is determined by the clinical course and by the precise vascular anatomy of the lesion as defined by MR imaging and spinal angiography. Treatment may involve endovascular embolization, surgery, or both.8,9,12 We report on the successful endovascular embolization of a large spinal MAVF in an asymptomatic 3-year-old boy with HHT.
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Pedigree:

![Pedigree Diagram]

**Fig. 1.** Diagram of the patient’s family pedigree. The proband is indicated by the arrow in the lower left corner. NL = normal.

**Case Report**

*History and Examination.* This healthy 3-year-old boy with a family history of HHT was evaluated at 18 months of age for a punctate cutaneous telangiectasia on the left pinna. There was no history of epistaxis or melena. Results of physical and neurological examinations were normal, and no cranial or abdominal bruits were present.

*Family History.* Hereditary hemorrhagic telangiectasia was diagnosed in the proband’s 32-year-old mother during adulthood after an episode of tunnel vision and expressive aphasia. A ventilation perfusion scan revealed a right-to-left shunt, and she subsequently underwent embolization of multiple pulmonary AVMs. Screening MR imaging and subsequent angiography procedures in the proband’s younger brother revealed a small, asymptomatic pial AVM in the right posterior temporal and occipital lobe, which was shown to be thrombosed on angiography and follow-up imaging. His maternal grandfather had undergone partial pulmonary lobectomies during adolescence as well as embolization of a large pulmonary AVM in middle age. His maternal uncle died at 6 months of age of spontaneous intracranial hemorrhage. His maternal great-grandfather died at 38 years of age of “pneumonia” (Fig. 1).

*Evaluation and Treatment.* Because of the severity of the presenting manifestations of HHT in several affected family members, imaging screens of the proband were undertaken. Cerebral angiography and MR imaging revealed small low-flow pial AVMs in the right posterior medial frontal lobe and in the right occipital lobe. Chest and abdominal CT scanning at 23 months of age detected no pulmonary or hepatic AVM but incidentally revealed a probable AVM in the thoracic spinal canal.

Spinal canal MR imaging revealed an intramedullary varix with surrounding spinal cord edema (Fig. 2). Subsequent spinal angiography, including selective angiograms of the right L-1 and L-2 spinal artery pedicles and the left L-1 spinal artery pedicle (Fig. 3), documented a large MAVF fed by the posterior spinal arteries of T-12 and L-1 on the left and L-2 on the right. These arteries coalesced into a single, 1-cm long artery that then entered the AVF into the large intramedullary venous varix at T-11. Venous drainage was via numerous perimedullary veins, with congestion of the surrounding spinal cord. The remaining cervical, thoracic, and lumbar spinal arteries appeared normal, with the anterior spinal artery arising from the right T-11 intercostal artery. The large size of the intrinsic varix and extensive cord edema were believed to represent a high risk of sudden neurological deterioration. The causative AVF appeared to be definitively treatable by using an endovascular approach. Thus, despite the asymptomatic presentation of this MAVF, endovascular treatment was recommended.

*Embolization Procedure.* At 3.5 years of age, the patient underwent transarterial embolization of the MAVF after induction of general anesthesia. Vasospasm was encountered during the attempt to access the left L-1 lumbar artery. Therefore, a flow-directed microcatheter was navigated via the right L-2 spinal artery. The catheter was navigated into the corresponding posterior spinal artery to the level of the AVF. Superselective angiography revealed filling of the fistula and no normal spinal branches. Embolization at this location was performed with a 3:1 mixture of N-butyl cyanoacrylate and ethiodol, achieving angiographically successful obliteration of the fistula (Fig. 3).

**Results**

Postembolization selective angiography of the left L-1
spinal artery demonstrated no residual filling of the varix via any of the previously identified feeding vessels, indicative of complete obliteration of the MAVF. Patency of the posterior spinal artery branch arising from the right T-11 pedicle was demonstrated, confirming intact blood supply to the conus medullaris and lower thoracic spinal cord after embolization via the normal artery ofAdamkiewicz and the anterior spinal artery. The venous phase of the angiogram demonstrated resolution of venous congestion. The patient was closely monitored for lower-extremity neurological function in a pediatric intensive care unit with ongoing neurosurgical consultation (in the event of neurological deterioration from variceal swelling that might require urgent operative exploration). No loss of neurological function was detected.

Postembolization axial and sagittal T2-weighted MR images (Fig. 2 lower left) revealed reduction in size of the venous varix and partial resolution of the associated spinal cord edema. There were no neurological or local complications of the angiographic procedure. The patient’s lower-extremity and sphincter function and neurological examination remained normal at 10 months after the embolization. A sagittal T2-weighted MR image obtained 10 months after the embolization (Fig. 2 lower right) revealed complete resolution of T2 hyperintensity in the spinal cord. No abnormal perimedullary flow voids were apparent. Hemosiderin deposits were evident within the residual cavity of the venous varix.

Discussion

The true incidence of both symptomatic and silent spinal cord AVFs in patients with HHT is unknown. In a review of 200 such patients with neurological symptoms, 36% had vascular lesions in the CNS, including 8% with spinal cord AVFs.13 Children with HHT and spinal cord AVFs presented most commonly with neurological deficits resulting from hemorrhage or from spinal cord venous hypertension. In a recent study of 155 patients, specific angioarchitectural characteristics of arteriovenous shunts of the spinal cord did not correlate with predisposition to hemorrhage.10 Progressive neurological deficits may result from congestion of the ectatic perimedullary venous channels.10 Although our young patient with HHT was asymptomatic, the presence of a large venous varix and spinal cord edema suggested high risk for neurological deficit and potential hemorrhage. Endovascular embolization of the MAVF allowed us to successfully obliterate this potentially morbid lesion without neurological deficit or an extensive operative procedure.

Conclusions

Endovascular treatment of spinal cord MAVFs can be performed successfully in very young children with HHT. Detailed selective angiography is essential to define the precise vascular anatomy of the MAVF. Interdisciplinary neurosurgical and interventional radiological consultation are essential for diagnostic and therapeutic management. Magnetic resonance imaging of the CNS should be considered to screen children with HHT who have a family history of neurological morbidity to reduce the risk of severe
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neurological injury should this disease present in the CNS. Large, prospective studies of families with HHT will be necessary to more accurately determine the true incidence of spinal cord A VFs in patients with HHT and to formulate evidence-based screening guidelines. Conversely, the discovery of a spinal cord MAVF in a young child is often associated with the presence of HHT and should prompt diagnostic consideration of HHT and clinical genetic evaluation of the family, because the characteristic presenting combination of mucocutaneous telangiectasia and epistaxis may not yet have developed.9

References

Fig. 3. Preembolization and postembolization angiography images of the right L-2 (A, D, and E) and left L-1 (B and C) spinal artery pedicles. The right L-2 (A) and left L-1 (B and C) feeding posterior spinal radiculomedullary arteries supply the stenotic perimedullary AVF adjacent to the large varix at T-11, which then drains into dilated and congested perimedullary veins on the later venous phase image (C). The left L-2 spinal artery (D) has complete occlusion of the fistula with preservation of the cord vascular supply from the posterior and anterior spinal radiculomedullary branches and a normal artery of Adamkiewicz, arising from the right T-11 intercostal artery (E).