Schimmelpenning syndrome is a rare neurocutaneous disease characterized by an obligatory craniofacial nevus sebaceous in association with seizure, developmental delay, and ocular or skeletal disorders. Extracutaneous features of this syndrome include cardiovascular anomalies, vascular malformations, urological defects, vitamin D–resistant rickets, benign tumors, numerous malignancies, and intraoral lesions. Cerebral abnormalities are also reported in a large percentage of patients with Schimmelpenning syndrome. These patients typically exhibit hemimegalencephaly and become symptomatic with seizures. We report the first case of a spinal AVM associated with Schimmelpenning syndrome.

Case Report

History. This 13-year-old girl with a history of Schimmelpenning syndrome presented to another facility with rapid onset of upper-extremity paresis and lower-extremity monoplegia. Imaging, including a cervical MRI study, revealed hemorrhage associated with a cervical AVM.

The patient’s medical history was notable for nevus sebaceous of the left ear, cheek, and anterior neck. The lesions involving her ear and neck had been resected. She had already undergone repair of coarctation of the aorta at the age of 2 months, resection of an epibulbar dermoid, and repair of cutis aplasia. She had also received a diagnosis of dysplastic left kidney; discrepancy in the length of her left leg compared with her right; and multiple osseous lesions of her left shoulder, femur, and pelvis that were radiographically consistent with enchondromas. An MRI study of the brain performed at birth had shown a left temporal arachnoid cyst and right hemimegalencephaly (Fig. 1), although she did not suffer from seizures or significant developmental delay, and other features of hemimegalencephaly such as cortical dysplasia and multiple layers of cortex are lacking.

One month before the patient’s acute presentation, she underwent surveillance MRI of the brain, which demonstrated stability of the arachnoid cyst and hemimegalencephaly. Abnormal flow voids in the region of the cervical spinal cord prompted dedicated MRI of the cervical spine. This study showed a large spinal AVM extending from the cervicomedullary junction to C-7 (Fig. 2). The
AVM consisted of intramedullary, intradural extramedullary, and osseous (C-5 body) components. She was scheduled for neurosurgical evaluation when she suffered the hemorrhagic event.

Spinal angiography performed at another institution showed a flow-related aneurysm off a branch of the right thyrocervical trunk. The aneurysm, which was thought to be the site of hemorrhage, was successfully embolized using coils (Fig. 3). The patient was discharged to a rehabilitation facility, where she partially recovered her strength. She was referred to our facility 4 months after the hemorrhage for further evaluation of the AVM.

Examination. On our initial evaluation the patient had antigravity strength in her proximal upper extremities bilaterally and functional strength in her hands. She was nonambulatory, with minimal movement in her bilateral lower extremities. She was scheduled for staged embolization followed by resection of the lesion, with the primary objective of spinal cord decompression.

Operation. At our institution, the embolization was performed using Onyx (ev3 Inc.) and N-butyl cyanoacrylate (Cordis Neurovascular) in 2 stages via right femoral artery and combined right femoral and left brachial artery approaches, due to an absence of the origin of the left subclavian from the arch (Fig. 4A and B). Resection proceeded through a C-1 laminectomy and C2–6 laminoplasty and standard microscopic dissection and resection (Fig. 5). Postoperative angiography showed a small amount of intramedullary residual AVM and an osseous portion involving the C-5 vertebral body (Fig. 4C and D).

Postoperative Course. The patient tolerated all interventions well. After the resection, she suffered a transient neurological decline. However, by the 3rd postoperative day her neurological function recovered to her preoperative baseline. During this period she was monitored in the ICU and her blood pressure was strictly controlled. Subsequently, she was discharged to a rehabilitation facility.

Discussion

Schimmelpenning syndrome is a rare neurocutaneous disorder defined by a craniofacial nevus sebaceous associated with seizure, developmental delay, and ocular or skeletal disorder.1,4 In 1957 Schimmelpenning12 described a patient with nevus sebaceous, seizures, and ocular abnormalities. In 1962, independent of this first report, Feuerstein and Mims2 reported 2 patients with nevus sebaceous, seizures, and mental retardation. Solomon et al.14 later coined the term “epidermal nevus syndrome” after reviewing 60 cases of patients with epidermal nevi and accompanying extracutaneous abnormalities. Subsequently the literature has been muddied by the use of numerous names for the syndrome, including epidermal nevus syndrome, linear sebaceous syndrome, epidermal nevus syndrome of Solomon, organoid nevus phacomatosis, Schimmelpenning-Feuerstein-Mims syndrome, Jadassohn nevus phacomatosis, organoid nevus syndrome, nevus sebaceous syndrome, Feuerstein-Mims syndrome, linear nevus sebaceous syndrome, and Solomon syndrome. Recently, an attempt has been made to clarify the nomenclature by emphasizing the name “Schimmelpenning syndrome” as a single syndrome within a group of epidermal nevus syndromes.4

Various neurological manifestations of Schimmelpenning syndrome have been reported.1,9 With rates
ranging from 60% to 78%, seizures and developmental delay are the most common symptoms. Hemimegalencephaly and unilateral ventricular dilation are the most common radiographic findings. Our patient exhibited hemimegalencephaly but did not have seizures or significant developmental delay. Other CNS anomalies include porencephaly, focal cortical dysgenesis, pachygyria, heterotopic gray matter, optic glioma, and brain AVMs. Two cases of brain AVMs have previously been reported. It is not surprising that other regions of the CNS could be involved. In fact, a thoracic intraspinal hemorrhage causing paraplegia has previously been reported; however, angiography studies obtained in that patient did not identify a causative lesion.

Greene et al. carefully reviewed the literature and presented their own series of patients with Schimmelpen-
Spinal AVM with Schimmelpenning syndrome

ning syndrome and associated vascular anomalies. These vascular anomalies, most frequently venous malformations and arterial malformations of the systemic vasculature, were present in 33% of their patients and in 13% of patients in the literature. Other malformations observed included coarctation of the aorta, aberrant dural sinuses, a cervical carotid artery malformation, and a leptomeningeal hemangioma. Based on the presence of these various lesions, Greene et al. concluded that patients with Schimmelpenning syndrome are at increased risk of vascular anomalies. Indeed, our patient had undergone repair of her coarctation of the aorta at 2 months of age.

The best-known neurocutaneous syndrome involving spinal AVMs is Cobb syndrome or cutanomeningospinal angiomatosis, which is defined as the presence of a cutaneous nevus and a spinal AVM in the same dermatome.6,13 Similarly, Sturge-Weber syndrome involves angiomas of the leptomeninges, skin, and face.13 Some authors have suggested an association between Schimmelpenning syndrome and Sturge-Weber because of the presence of cerebral vascular malformations, the cutaneous findings typically involving the face, and the lack of heritability in both syndromes. However, this association is unlikely because the characteristic facial capillary malformation in Sturge-Weber is absent in Schimmelpenning, and the cerebral vascular malformations are markedly different in both frequency and type.3

Schimmelpenning syndrome is a rare neurocutaneous syndrome with a propensity for both systemic and cerebral vascular anomalies. We report the first case of a patient with this syndrome who also had a concomitant large spinal AVM. Neurosurgeons should be aware of this rare phacomatosis and of the various neurological pathologic entities associated with this diagnosis. A low threshold for imaging the neuraxis should be exercised in these patients.

Disclosure

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

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![Fig. 5](image-url) Intraoperative photographs demonstrating the AVM before (A), during (B), and at the conclusion (C) of resection. Used with permission from Barrow Neurological Institute.
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