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### Sturge-Weber Syndrome Variant With Atypical Intracranial Findings: Case Report

#### ABSTRACT

Sturge-Weber syndrome is characterized by a facial port-wine nevus, leptomeningeal angiomatosis, and glaucoma; it is commonly complicated by epilepsy and hemiparesis. We present a patient with a head and neck port-wine nevus, glaucoma, abnormalities of the intracranial deep veins, and untreated communicating hydrocephalus. The patient lacks any radiologic or clinical evidence of cerebral leptomeningeal angiomatosis. Considering that intracranial venous anomalies also are likely compatible with the embryologic explanation of Sturge-Weber syndrome, this child can serve as an unusual example of Sturge-Weber syndrome type II. (*J Child Neurol* 2006;21:155–157; DOI 10.2310/7010.2006.00026).

The criteria for Sturge-Weber syndrome, also known as encephalofacial angiomatosis, have broadened significantly since its earliest descriptions. In 1860, Schirmer described a patient with a facial port-wine stain who had an associated finding of buphthalmos with no neurologic deficit noted.<sup>1</sup> Nineteen years later, Sturge described another patient with a facial port-wine stain with buphthalmos who was hemiparetic.<sup>1</sup> Sturge postulated the possibility of a vascular nevus in the brain similar to the facial angioma.<sup>1</sup> In 1929, Weber described the associated intracranial abnormalities with calcifications.<sup>1</sup> Currently, the most common features of this syndrome include a facial port-wine nevus, leptomeningeal angioma, and glaucoma. However, patients can have varying degrees of or a complete absence of one of these characteristic findings.<sup>2</sup> Roach proposed a classification system to address this broad spectrum of Sturge-Weber syndrome presentations.<sup>2</sup> The type I classification has both facial and leptomeningeal angiomas. Type II has a facial angioma without central nervous system leptomeningeal angiomatosis. Type III has isolated leptomeningeal angiomatosis without a facial port-wine nevus. We report the case of a boy with a classic facial port-wine nevus, glaucoma, and developmental malformations of the intracranial deep veins with communicating hydrocephalus but without leptomeningeal angiomatosis. The intracranial venous abnormalities are additional support for the diagnosis of Sturge-Weber syndrome type II in this patient.

#### Case Report

This 12-year-old Hispanic boy was initially evaluated in 1994 in Honduras, Central America. When first seen at the age of 2 years, he had a history of a normal gestation, a normal vaginal delivery, and normal development. There was no history of seizures. He had an unremarkable family history. A cutaneous port-wine nevus was apparent involving the distributions of all three divisions of the left trigeminal nerve and of the second and third divisions of the right trigeminal nerve and adjacent neck (Figure 1). His head circumference was 58 cm. The funduscopic examination was normal. No focal or generalized neurologic deficits were detected. When reevaluated

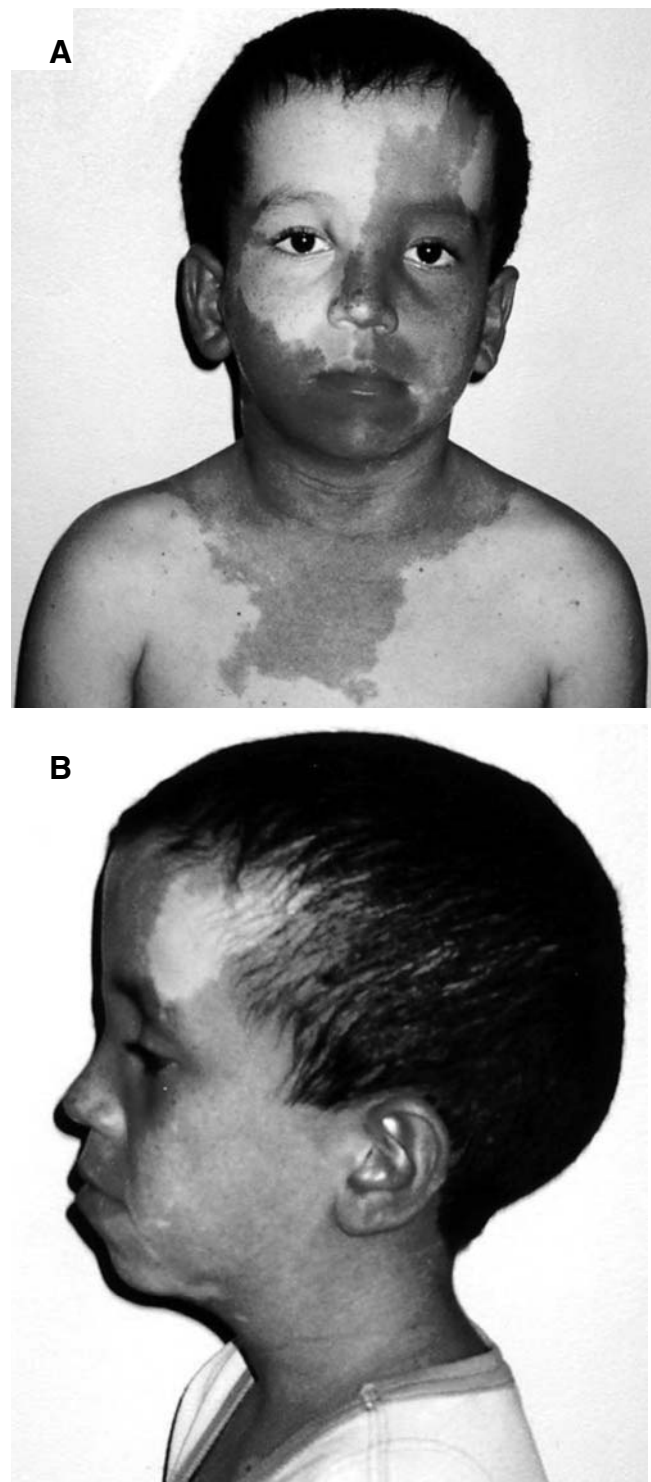


Figure 1. A, B, Patient with port-wine nevus in the distribution of all three divisions of the left trigeminal nerve and of the second and third divisions of the right trigeminal nerve and neck. Macrocephaly and nasal bridge fullness are also present.

ated at 3 years of age, his neurologic development was normal, with no history of headaches or decreased vision. His head circumference was 59 cm. There was bluish puffiness around the bridge of his nose and eyes. Brain magnetic resonance imaging (MRI) with contrast demonstrated diffuse symmetric ventricular enlargement without evidence of obstruction of ventricular cerebrospinal fluid drainage pathways, focal anatomic defects, or

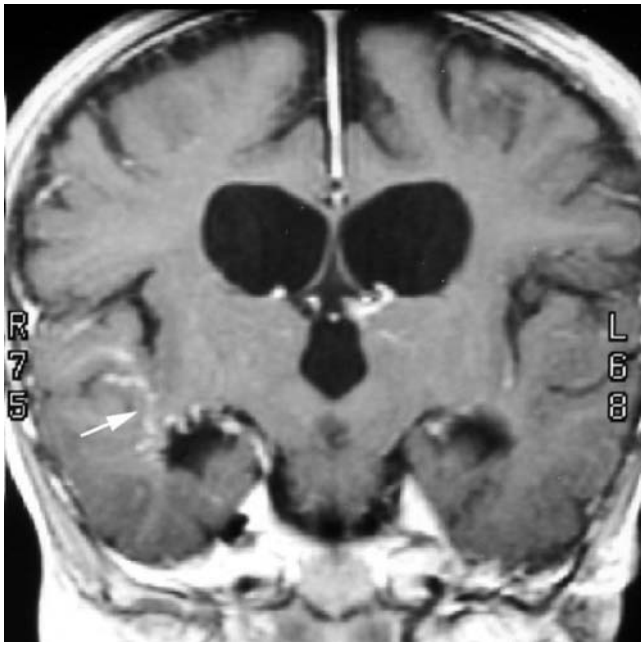


Figure 2. T<sub>1</sub>-weighted coronal gadolinium DTPA-enhanced brain MRI demonstrates symmetric lateral ventricular dilatation but no evidence of obstruction of CSF drainage at the level of the foramina of Monro or of enhancing leptomeningeal angiomatosis. Note an abnormally prominent deep vein (arrow) reflecting the altered venous drainage in this patient.

leptomeningeal angiomatosis (Figure 2). This coronal T<sub>1</sub>-weighted image also demonstrates some small enhancing internal cerebral veins in the roof of the third ventricle. The patient was diagnosed with communicating hydrocephalus.

At 4 years of age, follow-up evaluation revealed an unchanged facial port-wine nevus, no seizures or headaches, and no neurologic deficits or developmental delays. The head circumference was 60 cm. Glaucoma was suspected in the left eye. Arrangements were made for a pediatric neurologic/neurosurgical and ophthalmologic evaluation in the United States. He was found to have glaucoma in his left eye, which was treated. He underwent brain computed tomography and cerebral angiographic studies to evaluate the communicating hydrocephalus and suspected abnormal venous drainage in the area of his eyes and nasal bridge (Figure 3). These studies revealed an abnormal symmetric enlargement of the lateral and third ventricles consistent with communicating hydrocephalus on his previous brain MRI and a dilated right greater than left superior ophthalmic vein with retrograde venous flow draining through the angular and facial veins. This venous blood had originated from the middle cerebral veins and flowed into the cavernous sinus from the sphenoid sinus and then retrograde through the ophthalmic veins. Additional findings showed cortical venous drainage passing into the superior sagittal sinus and to the torcular Herophili, as well as from the veins of Labbé into the torcular. Venous drainage from the torcular passed into the occipital sinus and marginal sinuses and then out through the foramen magnum. The deeper parts of the brain appeared to drain both transcortically into the cavernous sinuses and into the inferior sagittal sinus and through a persistent falcial vein to the torcular and out through the foramen magnum. There was no evidence of abnormally rapid arteriovenous shunting to suggest an arteriovenous fistula involving the cavernous sinus. The straight sinus, vein of Galen, sigmoid sinuses, basal veins of Rosenthal, internal cerebral veins, and thalamostriate veins were not visualized on a conventional arteriogram (see Figure 3A). Some internal cerebral veins were noted on the previous brain MRI (see Figure 2). No abnormal cortical calcifications were present. No ventriculoperitoneal shunt was placed for the child's hydrocephalus at the time because of suspected mild

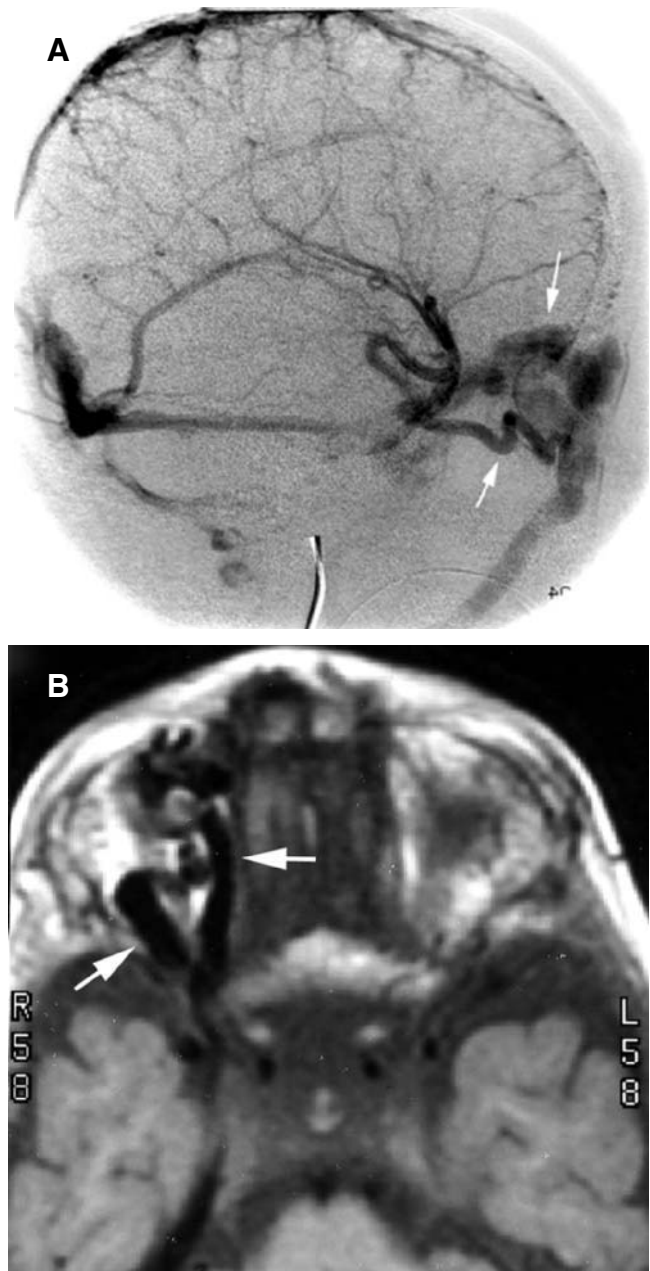


Figure 3. A, Venous phase from right carotid arteriogram (lateral projection) demonstrates absence of deep (Galenic) veins and abnormal retrograde venous drainage via markedly enlarged orbital veins (arrows). B, Axial T<sub>1</sub> image demonstrates vascular signal voids within abnormally enlarged right orbital veins (arrows).

diffuse pial angiomatosis with the associated risk of intraoperative hemorrhage and early evidence of compensating hydrocephalus.

By 7 years, the head circumference was 61.25 cm. The port-wine nevi and venous drainage through the ophthalmic and facial veins were clinically unchanged. His left eye glaucoma was well controlled with medication. The patient exhibited above-average psychomotor development for age and had no visual abnormalities, headaches, or seizures. At 11 years, his head circumference was 62 cm. No neurologic abnormalities or developmental delays were present. At 12 years old, his head circumference plateaued at 62 cm. His glaucoma remains controlled with timolol maleate drops. He has not had any significant headaches, visual complaints, or seizures. His psychomotor development remains above average for age.

## Discussion

The characteristic findings seen in Sturge-Weber syndrome are thought to arise from an embryologic malformation affecting the vessels that supply the face, eye, and brain.<sup>3</sup> The port-wine nevus in our patient affected the left eye and both sides of his face and neck. Port-wine nevi are caused by a malformation of small veins and capillaries beneath the surface of the skin. It is thought that this vascular malformation originates during the first trimester of gestational development when the primitive cephalic venous plexus fails to regress.<sup>3</sup> Fewer than 50% of children with port-wine nevi have pial angiomas with the corresponding neurologic complications of Sturge-Weber syndrome.<sup>1</sup> Some children can lack a port-wine nevus and still have pial angiomas.<sup>2</sup>

Commonly in Sturge-Weber syndrome, there is dilation of the intracranial deep veins that occurs because of malformation or thrombosis of the smaller cortical veins.<sup>4</sup> In our patient, there is almost complete absence of the deep venous system with shunting of blood through the cavernous sinus into the ophthalmic veins to the facial veins. Deep venous anomalies are not unusual in Sturge-Weber syndrome, and progressive venous sinus occlusion has been documented with magnetic resonance venographic techniques.<sup>5-7</sup> What is rare is that the dominant venous drainage from the intracranial venous anomalies is transorbital while at the same time there is apparent absence of a pial angioma. It is suspected in our patient that there was a developmental malformation resulting in the congenital absence or subsequent occlusion of multiple deep veins with redirection of intracranial venous flow through the orbital veins.

Communicating hydrocephalus occurs when the cerebrospinal fluid is obstructed at the level of the arachnoid villi.<sup>8</sup> The probable origin of our patient's communicating hydrocephalus is from the increased intracranial venous pressure resulting from his congenital intracranial venous malformations. The elevated venous pressure impedes normal resorption of cerebrospinal fluid across the arachnoid villi into the venous sinuses. By developing the hydrocephalus as an infant when his sutures had not yet closed, his skull was able to expand in response to the increased cerebrospinal fluid long after 2 years of age.<sup>8</sup> Because of this expansion and development of megalencephaly as a compensation for increased intracranial pressure, he has avoided any neurologic complications from compression of brain tissue. The head circumference has plateaued, and his sutures have closed to palpation.

Schirmer's first reported patient with a facial port-wine nevus who had buphthalmos did not have a neurologic deficit.<sup>1</sup> In Sturge-Weber syndrome, the glaucomatous eye is commonly on the ipsilateral side of the child's face with the port-wine nevus in the first division of cranial nerve V ( $V_1$ ) distribution.<sup>9,10</sup> The glaucoma is thought to result from an episcleral hemangioma causing elevated episcleral venous pressure.<sup>9</sup> Our patient had a port-wine nevus in the cranial nerve  $V_1$  and  $V_2$  trigeminal distributions on the left side of his face and had left eye glaucoma, which has been well controlled with medical treatment.

The lack of any radiologic evidence or clinical neurologic findings typical of pial angiomas raises doubt if our patient should be considered as having Sturge-Weber syndrome even though he has a classic facial port-wine nevus and glaucoma.<sup>11</sup> The congenital absence of most of the intracranial deep draining veins with anomalous venous drainage raises the question of whether this is another variant in Sturge-Weber syndrome or a different syndrome altogether. Our diagnosis of Sturge-Weber syndrome type II is primarily based on the Roach classification, but this patient might be an example of a wider group of embryologic cranial venous abnormalities of which Sturge-Weber syndrome is just a part.

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## Impaired Motor Control in Patients With Benign Focal Epilepsy of Childhood

### ABSTRACT

Upper extremity motor function was quantitatively assessed in 6 children (age 7-11 years) treated with antiepileptic drugs for benign focal epilepsies of childhood and compared with that of 30 age-matched normal children. Both motor performance and adaptation to perturbing mechanical constraints imposed by a robotic device were significantly impaired in children with benign focal epilepsies of childhood. Our findings thus question whether certain "benign" epilepsies are truly benign and whether pharma-