CASE REPORT

Sturge–Weber syndrome with cerebellar involvement

Syndrome de Sturge-Weber avec atteinte cérébelleuse

M. Smith Pearl\textsuperscript{a}, W.M.A. Abdalla\textsuperscript{b}, D.D.M. Lin\textsuperscript{c}, A.M. Comi\textsuperscript{d}, E. Boltshauser\textsuperscript{e}, P. Gailloud\textsuperscript{a}, T.A.G.M. Huisman\textsuperscript{b,}∗

\textsuperscript{a} Divisions of Interventional Neuroradiology, Johns Hopkins Hospital, Baltimore, USA
\textsuperscript{b} Division Pediatric Radiology, Johns Hopkins Hospital, 600 North Wolfe Street, Nelson basement, B-173, MD 21287-0842 Baltimore, USA
\textsuperscript{c} Diagnostic Neuroradiology, Johns Hopkins Hospital, Baltimore, USA
\textsuperscript{d} Division of Neurology and Developmental Medicine, Department of Radiology and Radiological Science, Johns Hopkins Hospital, Kennedy Krieger Institute, Baltimore, USA
\textsuperscript{e} Division of Pediatric Neurology, University Children’s Hospital Zurich, Switzerland

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Summary
Sturge–Weber syndrome is a rare neurocutaneous disorder that typically presents with angiomata involving the face, ocular choroid and ipsilateral supratentorial leptomeninges. Posterior fossa involvement is extremely rare. We present two patients with simultaneous supra- and infratentorial involvement. Magnetic resonance imaging (MRI) and digital subtracted angiography (DSA) findings are discussed.

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MOTS CLÉS
Syndrome de Sturge-Weber ; IRM ; Angiographie numérisée soustraite

Résumé

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∗ Corresponding author.
E-mail address: thuisma1@jhmi.edu (T.A.G.M. Huisman).

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Introduction

Sturge–Weber syndrome (SWS), also known as encephalотrigeminal angiomatosis, is a rare neurocutaneous congenital disorder of uncertain inheritance [1] with an estimated incidence of one in 50,000 births [2]. SWS is believed to result from the absence of a normal superficial cerebral venous drainage [3].

SWS is characterized by angiomas involving the facial skin, typically in the distribution of the ophthalmic and/or maxillary nerve, ocular choroid and leptomeninges. Although diagnosed clinically by the association of a facial nevus with neurologic features such as seizures, hemiparesis, mental retardation or homonymous hemianopsia, intracranial involvement can only be determined by neuroimaging. Magnetic resonance imaging (MRI) is considered to be highly sensitive and specific. Imaging may reveal characteristic findings including pial and/or cortical enhancement, prominent deep draining intramedullary veins, choroid plexus hyperplasia, cortical calcifications and progressive cortical atrophy on follow-up [3–5]. Leptomeningeal angiomatosis (LA) typically involves one cerebral hemisphere, ipsilateral to the facial nevus. Up to 15% of children have bilateral cerebral involvement/disease [6]. Involvement of the cerebellum is extremely rare. We present two cases of SWS with diffuse LA involving the supra- and infratentorial brain.

Case reports

The first patient is a nine-year-old boy presenting with a large port-wine birthmark involving the left face, neck, upper chest and shoulder; headaches; seizures and left eye glaucoma. MRI showed an ipsilateral occipital and temporal LA combined with a mild atrophy. The left choroid plexus was enlarged. Dilated vessels were seen along the left middle cerebellar peduncle and trigonium of the lateral ventricle (Fig. 1A). To rule out a possible arteriovenous malformation, digital subtraction angiography (DSA) was performed. DSA revealed ‘‘tram track’’ curvilinear gyral calcifications and an abnormal blush along the medial aspect of the occipital and temporal lobes consistent with SWS (Fig. 1B). A paucity of normal cortical draining veins and several prominent collateral intramedullary-draining veins were documented. These findings corresponded with flow voids seen on MRI. Surprisingly, DSA also demonstrated cerebellar LA.

The second patient is a four-year-old boy with a port-wine birthmark involving the right face in the distribution of the ophthalmic and maxillary nerves. Initial computer tomography (CT) at 2.5 years of age revealed ipsilateral LA involving the right temporal and occipital lobes. At four years, the boy returned with a focal right-sided seizure with secondary generalization. MRI showed a mild cortical atrophy of the right temporal/parietal/occipital region (Fig. 2). In addition, a moderate involvement of the contralateral temporal/occipital cerebrum was seen. The choroid plexus within the right ventricle was significantly enlarged. Moreover, there was LA involving the left superior cerebellum without signs of cerebellar atrophy or calcifications.

Discussion

SWS is characterized by angiomas involving the face, ocular choroid and leptomeninges. SWS is believed to occur early in fetal life, between the fourth and eight week of gestation and results from failure of embryonic cortical veins to coalesce and develop. Consequently, the precirculatory plexus of primordial vessels persists. The close association between the ectoderm destined to become the skin of the face and the dorsal telencephalic vesicles programmed to become the occipital and parietotemporal lobes probably explains the concomitant manifestation of cutaneous facial nevus in the distribution of the trigeminal nerve, choroidal angioma and LA centered to the occipital lobes [3,4].

The abnormal superficial cortical venous drainage in SWS accounts for the main radiological features, which include increased leptomeningeal enhancement, gyral calcification, cortical atrophy and prominence of the deep venous system [3].

Leptomeningeal enhancement on MRI is considered the gold standard and most important criterion for the assessment/radiographic diagnosis of intracranial involvement in SWS [3,7–9]. The leptomeningeal vascular malformation is typically supratentorial and unilateral, predominantly overlying the occipital (95%), parietal (74%) and temporal (63%) regions. Involvement of the frontal region is less common [3,9]. Bilateral LA has been reported in 7.5 to 26% of patients [6,10,11]. Posterior fossa involvement in SWS is extremely rare, with only two cases reported in the literature [4,12]. The first case was a six-year-old boy with simultaneous involvement of the right tempo-occipital region and cerebellum. Clinical symptoms were not specific for cerebellar involvement [4]. The second case described cerebellar LA in a 23-year-old woman with extensive port-wine birthmarks all over the body and intracranial venous abnormalities characteristic of SWS, as well as additional anomalies suggesting a more complex developmental abnormality, possibly as a variant of SWS [13].

The presented two additional cases of simultaneous supra- and infratentorial LA can be explained by a defective development of embryonic cortical veins simultaneously involving the telencephalic and rhomboencephalic vesicle. The fact that the vascularization of the developing brain begins at the myelencephalon with an ascending sequential gradient through the metencephalon, mesencephalon, diencephalon and telencephalon would explain why in all presented cases of cerebellar involvement the cerebrum was also involved. Vice versa, this would also explain isolated involvement of the cerebrum. Simultaneous supra- and infratentorial involvement may represent a more severe end of the spectrum of SWS.

Our first case also showed that DSA might be helpful in examining the exact angioarchitecture and venous dynamics in SWS [4,7,14]. DSA may be indicated to rule out other vascular pathologies [3].

The diagnosis of the exact extent of LA is important because extensive LA is often associated with mental retardation and therapy refractory seizures [15]. In addition, surgical treatment is less likely an option. Interestingly, our patients had no clinical signs indicating cerebellar involvement.
Sturge–Weber syndrome with cerebellar involvement

Figure 1  Contrast enhanced axial T1-weighted MRI sequence (A) and conventional angiography (B) of patient 1. Contrast enhancing leptomeningeal angiomatosis is seen along the left occipital/temporal lobe. Digital subtracted angiography (DSA) shows an additional abnormal capillary blush along both cerebellar hemispheres.

Séquence IRM pondérée T1 avec injection (A) et angiographie cérébrale (B) du patient 1. L’angiomatose leptoméningée rehaussée par l’injection du produit de contraste est visible au niveau du lobe temporo-occipital gauche. L’angiographie numérisée soustraite montre une hypervascularisation capillaire anormale en regard des hémisphères cérébelleux.

Figure 2  Contrast enhanced T1-weighted coronal MRI of patient 2. Leptomeningeal angiomatosis is noted along the right occipital/temporal lobes and within the left cerebellar fissures consistent with simultaneous supra- and infratentorial leptomeningeal angiomatosis.

IRM pondérée T1 après injection dans le plan coronal du patient 2. L’angiomatose leptoméningée est visible en regard des lobes temporo-occipitaux et dans les fissures cérébelleuses traduisant une angiomatose leptoméningée simultanément supra- et infratentorielle.

Conclusion

LA in SWS is typically unilateral and supratentorial. Our cases showed that simultaneous involvement of the supra- and infratentorial brain might occur. Contrast enhanced MRI is the standard imaging modality. DSA is no longer utilized routinely. However, in rare cases such as illustrated in this report, DSA may better depict the nature and exact extent of vascular involvement and may facilitate our understanding of the pathophysiology of SWS.

References