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CASE REPORT

A rare presentation of Sturge-Weber syndrome: when a port-wine stain is not the key – case report

Miguel Labrusco[1a], Inês Martins[1b], Sara I. de Almeida[1c], Sofia Quintas[2d], and Paulo Oom[1e]*

[1]Paediatric Department; [2]Neurodevelopment Service, Paediatric Department. Hospital Beatriz Ângelo, Loures, Portugal

ORCID: [a]0000-0003-4979-0632; [b]0000-0003-4249-5542; [c]0000-0002-9332-122X; [d]0000-0001-8115-1852; [e]0000-0002-5329-8782

Abstract

Sturge-Weber syndrome (SWS) is a sporadic congenital vascular disorder which typically presents with a facial port-wine stain and capillary malformations affecting the brain and eye. We present the case of a 15-month-old boy admitted to the paediatric emergency department with right arm palsy and focal clonic seizure on the right side of the body, without a facial nevus. Brain MRI revealed leptomeningeal angiomatosis suggestive of SWS. Despite the absence of the characteristic facial port-wine stain, diagnosis of SWS remains clinical and must be suspected whenever there are focal seizures associated with focal neurological deficits. In this case report we describe the clinical presentation, imaging findings and management of this rare presentation of SWS. We are reporting this case as few patients have been described in the literature.

Keywords: Case report. Facial nevus. Focal seizures. Leptomeningeal angiomatosis. Sturge-Weber syndrome.

Uma apresentação rara do síndrome de Sturge-Weber: quando a mancha de vinho do porto não é a chave – relato de caso

Resumo

A Síndrome de Sturge-Weber (SWS) é um distúrbio vascular congénito esporádico, que se apresenta tipicamente por mancha facial de vinho do porto e malformações capilares com envolvimento cerebral e ocular. Apresentamos o caso de uma criança de 15 meses, do sexo masculino, admitida no Serviço de Urgência Pediátrica com paresia do membro superior direito e convulsão clónica focal no hemicorpo direito, sem mancha facial. A RMN cranioencefálica revelou angiomatose leptomeníngea sugestiva de SWS. Apesar da ausência da mancha de vinho do porto facial característica, o diagnóstico de SWS permanece clínico e deve ser suspeitado sempre que hajam convulsões focais associadas a défices neurológicos focais. Neste case report, descrevemos a apresentação clínica, imagiológica e abordagem desta rara forma de apresentação de SWS. Relatamos o caso atendendo ao escasso número de doentes descritos na literatura.

Palavras-chave: Angiomatose leptomeníngea. Case report. Convulsões focais. Mancha facial. Síndrome Sturge-Weber.

***Correspondence:**
Miguel Labrusco
E-mail: mptl@campus.ul.pt

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Keypoints

What is known

- SWS is a sporadic congenital vascular disorder which typically presents with a facial port-wine stain and capillary malformations affecting the brain and eye.
- SWS can be divided in 3 different types, depending on the affected region.
- Neurologic manifestations include seizures, focal neurological deficits, behavioural anomalies and progressive intellectual impairment of variable degree.

What is added

- Focal seizures and stroke-like events in infancy must raise suspicion of SWS, even in the absence of a facial port-wine stain.
- SWS without facial port-wine stain is rare and in these cases the diagnosis can be made upon typical brain imaging findings.
- Characteristic MRI findings include leptomeningeal enhancement, compatible with leptomeningeal angiomatosis.

Introduction

Sturge-Weber syndrome (SWS) is a sporadic, congenital, neurocutaneous disorder, typically presenting in infancy with a facial port-wine birthmark, ipsilateral leptomeningeal capillary malformations and vascular eye abnormalities[1]. The essential pathologic components are the trigeminal and leptomeningeal angiomatosis. Clinically, SWS can be divided in 3 different types: type 1, with the classic manifestation of facial port-wine stains and intracranial leptomeningeal angioma, type 2, with facial angioma without evidence of intracranial disease and type 3, with isolated leptomeningeal angioma[2]. Few reports describe this last variant, in which no facial nevus is observed[2-9]. In such cases, the presenting symptoms are typically seizures in up to 80% of patients, most commonly focal seizures[4,8]. The diagnosis depends on imaging findings such as leptomeningeal angiomatosis in brain magnetic resonance imaging (MRI) and intracranial calcifications in computed tomography (CT)[4].

In this case report, we present a rare case of a patient with leptomeningeal angiomatosis without an associated facial port-wine birthmark.

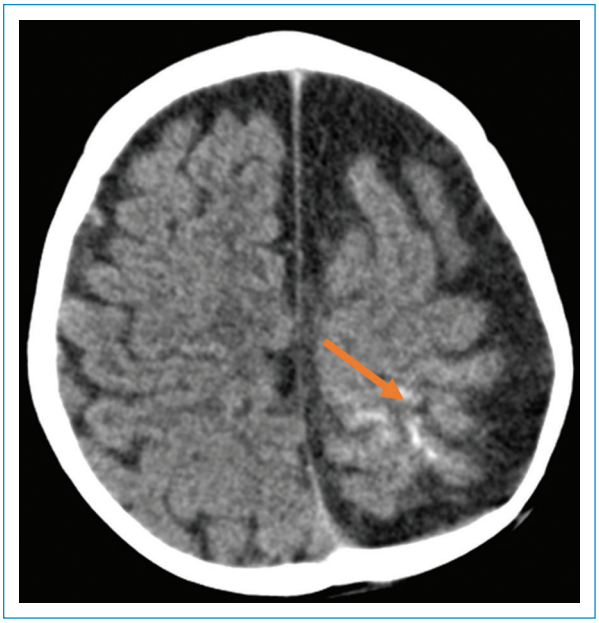

Figure 1. CT image with left cerebral atrophy with associated subcortical calcifications (arrow).

Case presentation

A previously healthy 15-month-old boy, with unremarkable family history, was admitted to the paediatric emergency department (PED) due to sudden onset right brachial palsy after waking up from a nap. Upon arrival at the PED, he presented clonic movements of the right hemibody with impaired awareness, without fever. The seizure was terminated after intravenous diazepam. In the post-ictal period, he maintained right hemiparesis and deep tendon hyperreflexia. There was no history of current or recent infection, trauma, medication or drug intoxication or other symptoms preceding this presentation. Physical exam revealed a borderline normal psychomotor development and preferential use of his left hand. Laboratory blood evaluation – hemogram, C-reactive protein and procalcitonin, had no alterations. Lumbar puncture was performed, and the cerebrospinal fluid (CSF) had no alterations.

Initial brain CT scan revealed left brain atrophy, most prominent on the fronto-parietal and temporal level, with associated subcortical calcifications on the fronto-temporal-occipital regions (Fig. 1).

He was admitted in the paediatric ward and started antiseizure medication (ASM) with levetiracetam as well as antibiotic and antiviral therapy with ceftriaxone and acyclovir while waiting for results from CSF cultures and virologic exam. One day after admission he started fever (tympanic temperature of 39.7°C every six hours), which progressively improved with sustained apyrexia

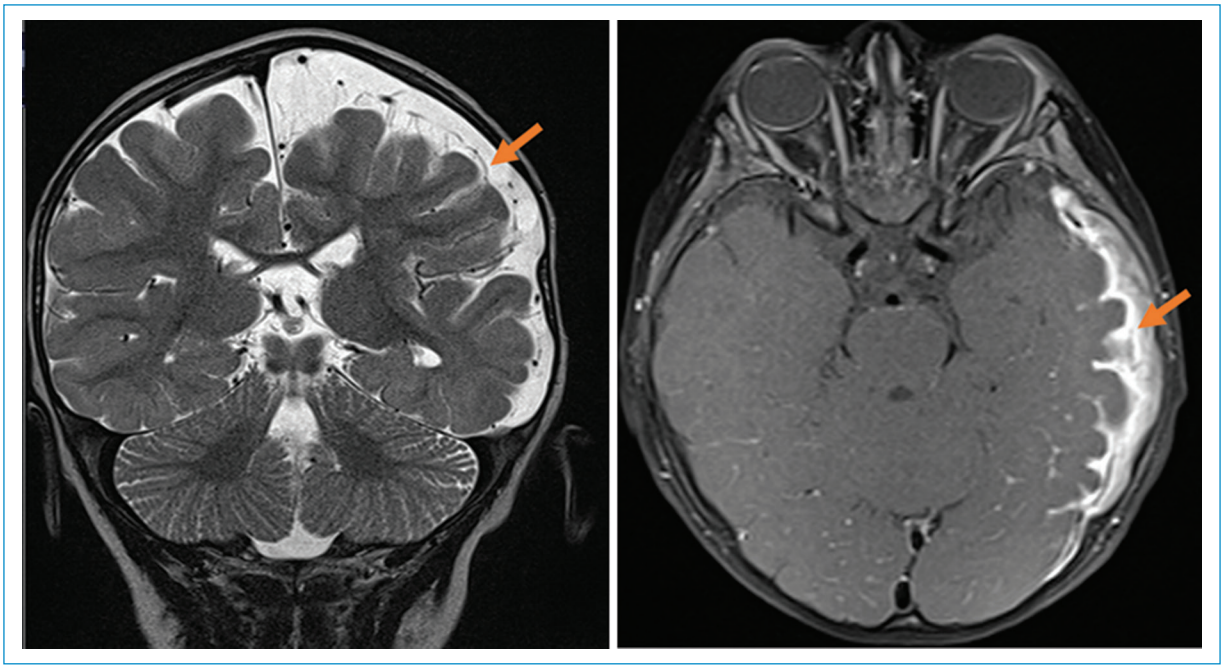

Figure 2. Coronal (left) and transverse (right) plane of MRI images with left cerebral atrophy, involving the frontoparietal and temporal regions. Marked leptomeningeal enhancement compatible with leptomeningeal angiomatosis (see arrow).

since day four of admission. Recent infection with Epstein-Barr virus and cytomegalovirus were ruled out. CSF and blood cultures were negative and so the antibiotic and antiviral were discontinued.

On the fourth day after admission, he had another focal clonic seizure, localized to the right hemibody, which stopped spontaneously after two minutes, followed by no clear post-ictal status or focal deficits. Subsequently, carbamazepine was added to levetiracetam showing a good response without significant side effects.

Electroencephalographic study disclosed an asymmetry of the basal activity (slower and minor amplitude on the left hemisphere, except for the occipital region), and no clear epileptiform activity. Brain MRI (Fig. 2) revealed left cerebral atrophy, involving the frontoparietal and temporal regions. Marked leptomeningeal enhancement compatible with leptomeningeal angiomatosis was suggestive of Sturge-Weber type III syndrome. These findings prompted the introduction of acetyl-salicylic acid (ASA). Ophthalmologic observation was normal.

At clinical discharge, he was apyretic and with no seizures for over 24 hours. He maintained ASM with carbamazepine and ASA. The child is undertaking occupational therapy with improved neurodevelopment.

Discussion

The diagnosis of Sturge-Weber syndrome should be elicited whenever the association of facial nevus and seizures are present[6]. However, rare variants of leptomeningeal angioma without the cutaneous lesion have been described[2-9].

In this case report, we presented a case of a previously healthy, 15-month-old male, with normal psychomotor development, admitted in the PED with sudden right brachial palsy, followed by homolateral focal motor seizures. Upon examination, there was no visible facial nevus. A CT scan reported left cerebral hemiatrophy associated with subcortical fronto-temporo-occipital calcifications, compatible with Sturge-Weber syndrome (Fig. 1). MRI study also revealed fronto-parietal leptomeningeal enhancement, compatible with leptomeningeal angiomatosis (Fig. 2).

Leptomeningeal angiomatosis can be confirmed histologically or by typical radiographic findings associated with suggestive clinical presentation[2]. Even though trigeminal and leptomeningeal angiomatosis are classic pathologic components of SWS, in our patient there was no evidence of trigeminal angiomatosis. The reason behind the absence of this facial nevus in such cases remains unclear.

Encephalofacial angiomatosis has been classified in three types, the present case is in accordance with type 3, in which an isolated leptomeningeal-brain angioma without facial nevus is present[10]. Leptomeningeal angioma is most commonly found in the occipital region. Case reports where this region is spared are rare[7].

SWS is caused by mosaic mutations in the GNAQ gene on fetal ectodermal tissues. Mutations occurring on earlier stages in embryogenesis may affect a greater variety of precursor cells, resulting in SWS[11]. The developmental process that leads to the meningeal angioma is postulated to result from an incomplete degeneration and subsequent maturation of the primitive cephalic venous plexus in the first trimester of development[2,7,12].

The slower blood flow caused by the angiomatosis can lead to stasis and consequent hypoxia, with subsequent cortical atrophy and changes in the vessel wall and ground substance that result in calcification, with characteristic hyperdensity found in neuroimaging, particularly in CT[2,4,12]. This finding should elicit other causes of cerebral calcification such as celiac disease, encephalitis, purulent meningitis, ossifying meningoencephalopathy or leukemia[4]. Leptomeningeal angiomas are readily identifiable with gadolinium-enhanced MRI, which is particularly useful for an early diagnosis, prior to the characteristic calcifications[7,13].

Non-specific findings can also be documented on electroencephalography, such as an asymmetric basal activity, with decreased voltage or slowing on the affected side[4], as was the case of our patient. When such electroencephalographic findings are associated with a typical facial nevus, this diagnosis should be suspected[2].

Neurologic manifestations include seizures, which are characteristically focal motor or generalized tonic-clonic, particularly in younger children, focal neurological deficits, including hypoxic-ischemic events and hemianopsia, behavioural anomalies and progressive intellectual impairment of variable degree[2-4].

The diagnosis of this condition is clinical. Genetic testing is available, however, the somatic, mosaic variants responsible for this pathology arise post-zygotically and only affected tissues will contain them, which limits the diagnostic efficiency[14,15].

There is no specific treatment for SWS[16], and management of neurologic manifestations is often difficult. Carbamazepine, oxcarbamazepine and levetiracetam are acceptable first-line options[17] for focal seizures.

Refractory epilepsy, particularly when associated with clinically significant hemiparesis, visual field loss and developmental delay, may benefit from surgical intervention such as hemispherectomy[18].

Low-dose aspirin beginning in infancy may also be beneficial, as antithrombotic therapy may prevent the progression of hypoxic-ischemic neuronal injury[19]. Some authors suggest the potential benefit of pre-symptomatic ASM treatment combined with aspirin, particularly in children with high risk for seizures[20], but these data still need validation.

The prognosis and clinical course are highly variable and it is unclear if the age of onset has influence, and some children experience intractable seizures, mental retardation, and recurrent stroke-like episodes[13].

This case highlights the variable clinical presentation of SWS, and the importance of its suspicion when presented with focal seizures and neurological deficits such as hemiparesis, even in the absence of facial nevus.

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Conflicts of interest

None.

Ethical disclosures

Protection of human and animal subjects. The authors declare that no experiments were performed on humans or animals for this study.

Confidentiality of data. The authors declare that they have followed the protocols of their work center on the publication of patient data.

Right to privacy and informed consent. The authors declare that no patient data appear in this article.

Use of artificial intelligence for generating text. The authors declare that they have not used any type of generative artificial intelligence for the writing of this manuscript, nor for the creation of images, graphics, tables, or their corresponding captions.

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