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Case report
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TYPE III ROACH SCALE STURGE-WEBER SYNDROME ASSOCIATED WITH AN EXTRACRANIAL LIPOMA - A CASE REPORT

VARIJANTA TIPA III STARDŽ-VEBEROVOG SINDROMA PO ROUČU UDRUŽENA SA EKSTRAKRANIJALNIM LIPOMOM – PRIKAZ SLUČAJA

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Summary

Introduction. Sturge-Weber syndrome is a neurocutaneous disorder characterized by neurological features such as headaches, developmental delay, mental retardation and seizures, facial anomalies such as port-wine stain and glaucoma which is the most common ocular manifestation. The main neuroimaging findings in patients with Sturge-Weber syndrome are leptomeningeal angiomas and corticopial calcifications associated with underlying cortical atrophy. The purpose of this report is to present a rare case of a patient with seizures whose magnetic resonance imaging findings suggested a Sturge-Weber syndrome variant. **Case Report.** We report a case of a 14-year-old boy with a two year history of well controlled generalized tonic clonic seizures with visual aura, who was admitted to our institution for neuroimaging examination. Neuropsychological testing showed normal cognitive and psychomotor development. Electroencephalography revealed unilateral runs of right occipital spikes with secondary generalization. Neuroimaging findings showed focal cerebral leptomeningeal enhancement in the right parasagittal occipital region associated with focal cortical atrophy, whereas susceptibility weighted imaging showed hypointense intracortical calcification and hyperplastic enhanced ipsilateral choroid plexus. The computed tomography confirmed cortical calcifications. Also, an overlying parieto-occipital subcutaneous lipoma was found in the innervation field of the ophthalmic nerve. **Conclusion.** Magnetic resonance imaging is the key imaging modality that confirmed the clinical suspicion of Sturge-Weber syndrome based on a physical and neurological examination. Neither magnetic resonance imaging nor clinical examination is sufficient for a correct diagnosis.

Key words: Sturge-Weber Syndrome; Lipoma; Seizures; Magnetic Resonance Imaging; Angiomas; Neuroimaging; Calcification; Diagnosis

Introduction

Leptomeningeal angiomas and corticopial calcifications are among the main features of Sturge-Weber syndrome (SWS). SWS is a neurocu-

Sažetak

Uvod. Stardž-Weberov sindrom je neurokutani poremećaj za koji su karakteristične neurološke tegobe kao što su: glavobolje, zaostajanje u razvoju, mentalna retardacija i epileptički napadi, anomalije lica kao što je mrlja boje *porto vina* i glaukom, kao najčešća očna manifestacija. Glavni neuroradiološki znaci kod pacijenata sa Stardž-Weberovim sindromom su leptomeningealna angiomatoza i kortikopijalne kalcifikacije udružene sa atrofijom moždanog parenhima ispod promene. Svrha ovog rada je prikaz retkog slučaja čiji neuroimidžing nalaz ukazuje na to da se radi o varijanti Stardž-Weberovog sindroma. **Prikaz slučaja.** Četrnaestogodišnji mladić sa dvogodišnjom istorijom, dobro kontrolisanih generalizovanih tonično-kloničnih napada sa vizualnom austom, primljen je u našu ustanovu radi neuroimidžing pregleda. Neuropsihološko testiranje je otkrilo normalan kognitivni i psihomotorni razvoj. Elektroencefalografski nalaz je prikazao unilateralnu patološku aktivnost okcipitalno desno sa sekundarnom generalizacijom. Nalaz na magnetno-rezonantnom imidžingu prikazao je leptomeningealno prebojavanje u okcipitalnoj regiji parasagitalno desno i kortikopijalne kalcifikacije, uz atrofiju parenhima ispod promene, kao i hipertrofiju istostranog horoidnog pleksusa bočne moždane komore. Pregled kompjuterizovanom tomografijom endokranijuma potvrdio je da se radi o kortikopijalnim kalcifikacijama. Dodatni nalaz je bio supkutani lipom parijetookcipitalno desno u regiji inervacije oftalmičkog nerva. **Zaključak.** Magnetna rezonancija je ključna radiološka metoda za potvrdu klinične sumnje na Stardž-Weberov sindrom, bazirane na kliničkom i neuropsihološkom ispitivanju.

Ključne reči: Stardž-Weberov sindrom; lipom; konvulzije; magnetna rezonanca; angiomatoza; neuroimidžing; kalcifikacije; dijagnoza

taneous disorder characterized by facial, ocular and cerebral vascular anomalies. Clinical presentation of SWS includes neurological features such as headaches, developmental delay, mental retardation and primarily seizures, as well as port-wine stain

Abbreviations

SWS	– Sturge-Weber syndrome
MRI	– Magnetic Resonance Imaging
T1W	– T1 weighted
T2W	– T2 weighted
FLAIR	– Fluid-attenuated inversion recovery
SWI	– Susceptibility weighted imaging
CT	– Computed tomography
MA	– Meningioangiomas
WISC	– Wechsler Intelligence Scale for Children

(PWS), mostly unilateral, in the innervation field of the ophthalmic nerve (V1), sometimes including the maxillary (V2) and mandibular (V3) nerves. It can also be bilateral; glaucoma is the most common ocular manifestation, probably due to vascular eye abnormalities. According to Roach, there are three types of SWS; type I includes facial and leptomeningeal angiomas with or without glaucoma; type II includes facial angiomas with or without glaucoma and no intracranial manifestations, and type III includes only leptomeningeal angiomas without facial and ocular lesions [1–4]. The purpose of this article is to present an unusual case of a young male patient with seizures whose neuroimaging findings suggested type III SWS associated with extracranial lipoma in the innervation field of the ophthalmic nerve and to discuss possible differential diagnosis of this unique finding.

Case Report

A 14-year-old boy, with a two-year history of well controlled generalized tonic clonic seizures with visual aura was admitted to our institution for neuroimaging examination. He was diagnosed with

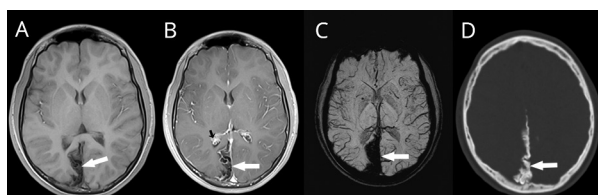


Figure 1. T1W before (A) and after (B) gadolinium administration in the axial plane; White arrows indicate leptomeningeal enhancement in the right parasagittal occipital region; Black arrow in B indicates hyperplastic enhanced ipsilateral choroid plexus; SWI (C) and CT (D) confirming the presence of “tram-like” corticopial calcifications in the same location

Slika 1. T1W pre (A) i nakon (B) primene gadolinijumskog kontrastnog sredstva u transverzalnoj ravni; bele strelice ukazuju na leptomeningealno postkontrastno pojačanje intenziteta signala okcipitalno parasagitalno desno, dok crna strelica (B) ukazuje na hiperplastični istostrani horoidni pleksus. SWI (C) i kompjuterizovana tomografija (D) potvrđuju prisustvo “šinastih” kortikopijalnih kalcifikacija na istoj lokaciji

Legend: SWI - Susceptibility weighted imaging

Legenda: SWI – imidžing magnetne susceptibilnosti

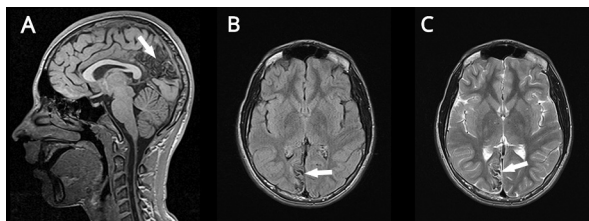


Figure 2. T1W in the sagittal plane (A), FLAIR (B) and T2W (C) images in the axial plane; White arrows indicate suspected abnormal leptomeningeal vessels in the right parasagittal, occipital region with focal atrophy of the surrounding cortex

Slika 2. T1W u sagitalnoj (A), FLAIR (B) i T2W (C) u transverzalnoj ravni. Bele strelice ukazuju na suspektu abnormalnu leptomeningealnu vaskulaturu u okcipitalnoj regiji parasagitalno desno, uz fokalnu atrofiju parenhima ispod promene

Legend: FLAIR – Fluid attenuated inversion recovery

Legenda: FLAIR – fluidni atenuirani inverzni oporavak

epilepsy and treated with valproic acid since the age of twelve. The seizure frequency was very low, with only two reported attacks per year, with similar semiology and duration. On admission, his neurological and physical states were unremarkable. Neuropsychological testing revealed normal cognitive and psychomotor development, with intelligence quotient (IQ) score of 126 on Wechsler Intelligence Scale for Children (WISC). Electroencephalography (EEG) was performed two days earlier and it revealed unilateral runs of right occipital spikes with secondary generalization. The Magnetic Resonance Imaging (MRI) showed a focal cerebral leptomeningeal enhancement in the right parasagittal occipital region (**Figure 2A**), associated with focal cortical atrophy (**Figures 1 and 2B**) and susceptibility weighted imaging (SWI) hypointense intracortical calcification (**Figure 2C**). As associated findings, hyperplastic enhanced ipsilateral choroid plexus and an overlying parietooccipital subcutaneous lipoma in the innervation field of ophthalmic nerve, were found (**Figure 3**). Non-enhanced computed tomography (CT) scans, performed subsequently, confirmed the presence of intracortical calcification (**Figure 2D**), revealing a (gyral) “tram-track” pattern, obscured by blooming susceptibility effect on SWI scans (**Figure 2C**). The ophthalmological examination, performed due to radiologically suspected SWS, did not reveal any ocular and retinal abnormality, including glaucoma.

Discussion

Even though the exact etiology of SWS remains unclear, it is proposed that the main pathophysiological mechanism in the affected regions of the brain is the absence of well-functioning superficial cortical venous system, thus blood is redirected centrally via medullary veins, resulting in venous hyperemia and hypertension [5].

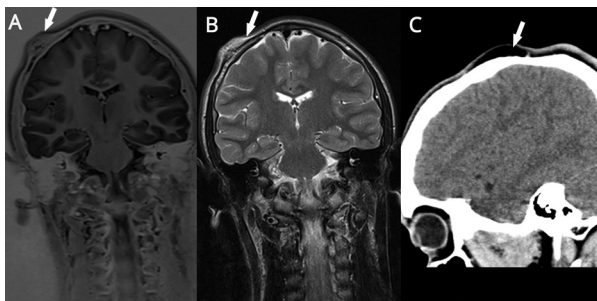


Figure 3. T1W (A) and T2W (B) in the coronal plane, and CT (C) in the sagittal plane; Arrows show a subcutaneous mass in the right parietooccipital region, with signal intensity and density of the fatty tissue, suggesting the presence of a subcutaneous lipoma

Slika 3. T1W (A) i T2W (B) u koronalnoj, kao i kompjuterizovana tomografija (C) u sagitalnoj ravni. Strelice ukazuju na supkutanu masu parijetookcipitalno desno, sa intenzitetom signala i denzitetom koji odgovara masnom tkivu, što ukazuje na prisustvo supkutanog lipoma

Neurological manifestations of SWS include seizures (75–90%), developmental delay (50–75%), headaches (40–60%) and hemiplegia (30%). The seizures are usually focal, arising from the areas of affected parenchyma, mostly parietooccipital. The early onset of seizures, larger unilateral lesions or bilateral disease, are risk factors for developing pharmacoresistant epilepsy and intellectual impairment [5–7]. Our patient had the first seizure attack at the age of twelve, and did not develop any other neurological or psychological disorders in the follow up years. On the contrary, his WISC score was 126, which puts him in the superior group. His mild neurological and normal neurocognitive status did not correlate with the pial angiomas and cortical atrophy extent, detected by MRI, affecting almost the whole medial surface of the right occipital lobe. These results prove that there is a big discrepancy between neuroimaging findings and clinical presentation in suspected cases of SWS variant, and neither clinical examination nor MRI alone is sufficient to make the correct diagnosis.

Regardless that the neuroimaging findings in our patient were highly suggestive of type III Roach variant of SWS, one should keep in mind, that similar imaging features could be present in other congenital pial angiomas related disorders and/or

angioproliferative diseases. First of all, the meningioangiomas (MA) should be considered in differential diagnosis, especially in pediatric patients and young adults with seizures [8]. MA is a rare benign lesion of unknown etiology, usually affecting cerebral cortex and leptomeninges, but can also involve the brain stem and thalamus [9]. It can occur sporadically or associated with neurofibromatosis type II (NF II) [10]. In our patient, the diagnosis of MA was ruled out, based on the lack of perilesional vasogenic edema in the surrounding brain parenchyma, which was reported as typical for MA.

The MRI features in our patient differ from previously reported cases of pediatric and young adults with SWS, in detected additional findings. Intracranial convexity lipoma, as a rare associative finding to focal leptomeningeal enhancement and corticopial calcifications, was described in unique case of Morana et al. The clinical presentation in their two non-related pediatric patients was similar to our patient [11]. To the best of our knowledge, extracranial subcutaneous lipoma, overlaying angiomas pial malformation in SWS has never been reported before. More interestingly, lipoma was completely within the innervation field of the right ophthalmic nerve. We believe that combined neuroimaging findings, such as pial angiomas and corticopial calcifications with atrophy of underlying brain parenchyma, in association with ipsilateral parietooccipital subcutaneous lipoma and seizures, without mental retardation and developmental delay, have not been reported yet, indicating a possibility of a rare manifestation of neurocutaneous disorder. The whole spectrum of clinical phenotypes associated with pial angiomas related disorders has still to be clearly defined.

Conclusion

Magnetic resonance imaging is one of the key imaging modalities that confirms the clinical suspicion of Sturge-Weber syndrome based on physical and neurological examinations. However, neither magnetic resonance imaging nor clinical examination alone is sufficient for a correct diagnosis of Sturge-Weber syndrome. In all pediatric and young adult patients, with clinical presentation of seizures, headaches, developmental delay or mental retardation and pial angiomas and cortical calcifications confirmed by magnetic resonance imaging, type III Sturge-Weber syndrome should be considered.

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