Berkala Ilmiah Kedokteran Duta Wacana (ISSN 2460-9684)
Volume 6, Number 1, 25 June 2021
http://dx.doi.org/10.21460/bikdw.v6i1.218

Case report

Sturge-Weber Syndrome in an epileptic child: a case report

Christofer Sathya Wijaya Budi Sarwono[1], Sutaryanu Darmoredjo[1,2]

1 Faculty of Medicine Universitas Kristen Duta Wacana, Yogyakarta, Indonesia
2 Department of Radiology Bethesda Hospital, Yogyakarta, Indonesia

Corresponding author — christofer.ito@gmail.com

Received: 12-10-2020 / Accepted: 27-05-2021

Abstract

Background Sturge Weber Syndrome (SWS) is a congenital neurocutaneous disorder with a facial capillary malformation, abnormal blood vessels of the brain, and abnormal blood vessels in the eye predisposing to glaucoma. The incidence of Sturge Weber Syndrome (SWS) is a rare entity that accounts about 3 of 1000 births with PWS, 5% of them suffer SWS.
Objective To describe the diagnosis of SWS in an epileptic children admitted to Bethesda Hospital, Yogyakarta
Case description A 5-years-old girl presented with a three-days history of muscle weakness of right upper and lower limb followed by headache, loss of consciousness, dizziness, with previous diagnosis of facial hemangioma without any symptoms like fever, nausea, vomit, and dysarthria. Head CT-Scan revealed a subcortical calcification at frontal lobe and left parietal with mild atrophy at bifrontalis lobes that support SWS diagnosis.
Conclusion Early detection of SWS among children with port-wine stain might improve patient prognosis.

Keywords: Sturge-Weber Syndrome, Port-Wine Stain, seizures, muscle weakness, Indonesia

Introduction

Sturge Weber Syndrome (SWS) is a rare congenital disorder characterised by port-wine stain, leptomeningeal lesion, and ocular haemangioma. Approximately 3 of 1000 births worldwide would present with port-wine stain, and 5% among them suffer from SWS. It occurs sporadically, not hereditary, and caused by a somatic mosaic mutation of the GNAQ gene. Three major clinical features of SWS are port wine stain in the first or second branch of trigeminal nerve, ipsilateral leptomeningeal capillary - venous malformation, and glaucoma.[1,2] Related to its rarity, SWS is scarcely studied, especially in Indonesia. This case report describes the SWS case in children with epileptic seizures at Bethesda Hospital, Yogyakarta.

Case Description

Patient Information

A 5-year-old female child was admitted to Bethesda Hospital, referred from a local hospital due to worsening muscle weakness of right upper and lower limb since three days before. In the ER, she presented with muscle weakness, headache, dizziness, and altered level of consciousness. There were no other systemic symptoms such as fever, nausea, or vomiting, or stroke-like sign such as dysarthria. Two days prior to admission, she experienced a partial seizure on the right part of the body for 2 minutes which followed by vomiting up to 5 times per day. There was no recurrency of seizure episode.

Her prenatal history was remarkable with regular antenatal visit and vaccination, although she was delivered through caesarean section due to pre-eclampsia and urinary tract infection. Her growth was normal, while she had delayed motoric and speech development.

She had prominent past history of epileptic seizures since 18 months of age, and took valproic acid 250 mg tablet per 12 hours every day for 4 years. In the past 4 years, she experienced more than 10 epileptic episodes at home, without further medical intervention. As her parent described, the seizure was tonic-clonic on the right body part, followed by decreased consciousness, and leave partial weaknesses of the right upper and lower limb. The history of fever, vomiting and dysarthria was denied. No history of similar epileptic seizures among her immediate family members.

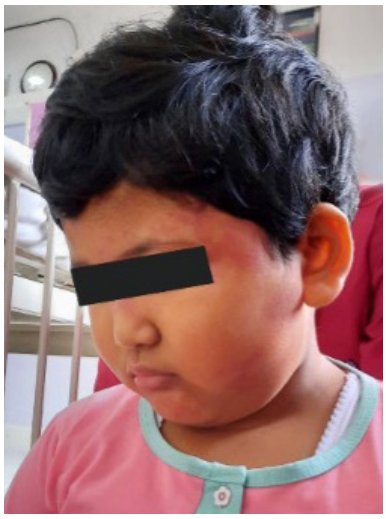

Figure 1. *Port-wine stain on the left face of the patient diagnosed as facial haemangioma*

Clinical findings

General physical examination was normal, with significant poor muscle strength on the right upper and lower limbs. Other neurological examination, including ophthalmology revealed no abnormalities. She had distinctive port-wine stain covering the left part of her face (Figure 1).

Diagnostic assessment

On the head CT scan, there was a subcortical calcification at frontal lobe and left parietal with mild atrophy at bifrontal lobes (Figure 2). The laboratory examination showed results in normal range. She was diagnosed with Sturge Weber Syndrome and complicated by Todd's paresis.

Therapeutic intervention

The patient was given intravenous phenytoin 75 mg per12 hour, oral piracetam 500 mg per 12 hour, and intravenous cefotaxime 500 mg per 8 hours. She also had oral valproic acid 250 mg/12 hour as maintenance treatment. The neurologist added oral aspirin 50 mg/24 hour. Additionally, she was prescribed a rehabilitation regiment by to improve her muscle strength and range of motion.

Discussion

Sturge Weber Syndrome is a sporadic neurocutaneous syndrome. It is characterized with three cardinal signs which are facial vascular malformation in ophthalmic nerve branches of trigeminal nerve (port-wine stain) and in ipsilateral leptomeninges which usually progressed to intracranial calcification, and glaucoma.[1]

SWS is strongly associated with a mutation of Q-class G protein alfa-subunit gene (GNAQ), which could activate mitogen-activated protein kinase (MAPK), and further induce cell proliferation and inhibit apoptosis especially in neurocutaneous cells. Mutation of somatic GNAQ might occurs during embryonic phase in the frontonasal prominence or during migration phase to neural crest where it will differentiate into leptomeningeal and choroid tissue.[1,2] This mutation affects vascular tissue which characterized with central venous malformation, ipsilateral leptomeningeal vascular malformation, absence of cortical superficial vein, and deep vein dilation. These conditions will induce absence of brain oxygen supply that leads to cerebral atrophy and calcification formation.[3]

Based on the cardinal signs, there are three types of SWS as described by Bodensteiner and Roach, which are type 1 (facial and leptomeningeal involvement, may involve glaucoma), type 2 (facial signs without intracranial involvement, small probability of glaucoma), and type 3 (intracranial lesion only, very low probability of glaucoma). Other less common signs and symptoms of SWS are seizure, hemiparesis or stroke-like events, behavioural problems, and intellectual disability.[3,4]

Main neuroimaging findings of SWS is the presence of the leptomeningeal capillary-venous malformation in brain MRI with gadolinium contrast. If it is not available, head CT scan usually could identify intracranial calcification. [1,3]

The aim of the treatment is mainly to control discomforting symptoms, such as seizure and headache. Routine follow up is needed to monitor the presence of stroke-like events, ophthalmological involvement, and behavioural problems. If possible, the facial capillary malformations might be treated with pulsed dye laser procedure. For some cases with severe neurological symptoms such as refractory seizures, hemispherectomy is advised as early as infancy period.[5,6]

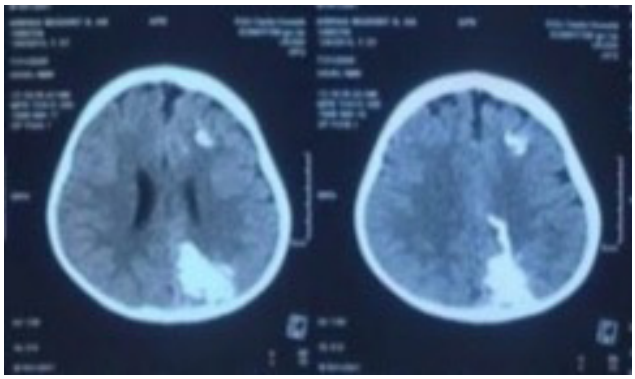

Figure 2. *Distinctive tram-line appearance on head CT-scan showing intracranial calcification*

Strength and limitation of clinical approach

The diagnosis of Sturge Weber syndrome in this patient was quite late compared to other cases[7–9], considering she suffered from refractory seizures for the past 4 year. Although the patient had hemiparesis in the previous seizure episodes, she was only prescribed a physical rehabilitation therapy at Bethesda hospital. This might be caused by its rarity, and lack of diagnostic and therapeutic experience of local hospital.

Compared to the literature[1,3], neuroimaging should be taken in earlier age to confirm the presence of intracranial lesions, especially when the patient presented with serious symptoms.

Since there is no definitive and curative treatment of SWS, after initial diagnosis, the patient could return to local hospital as the treatment is mainly to control her epileptic symptoms and physiotherapy.

Lesson learned

Sturge Weber Syndrome should be suspected in a child with port-wine stain and accompanying neurological symptoms as early as possible, mainly to control the symptoms and to monitor the occurrence of debilitating symptoms such as glaucoma and muscle weakness.

Conclusion

Sturge weber syndrome (SWS) is a rare congenital neurocutaneous disorder. It's rare occurrence, especially in Indonesia, is related to delayed diagnosis and poorer prognosis. Early neuroimaging detection in infant with port-wine stain and the presence of debilitating symptoms should improve the patient quality of life and prognosis.

Conflict of Interest and Funding Resource

The authors stated no conflict of interest.

Reference

1. Sahin M, Ullrich N, Srivastava S, Pinto A. Neurocutaneous Syndromes. In: Kliegman RM, Joseph W. St Geme, Nathan J. Blum, Samir S. Shah, Robert C. Tasker, Karen M. Wilson, editors. Nelson Textbook of Pediatrics. 21st ed. Philadelphia: Elsevier; 2020. p. 3140–50.
2. Varma R, Williams SD. Neurology. In: Zitelli BJ, McIntire SC, Nowalk AJ, editors. Zitelli and Davis' Atlas of Pediatric Physical Diagnosis. 7th ed. Philadelphia: Elsevier; 2018. p. 562–92.
3. Comi AM. Presentation, diagnosis, pathophysiology, and treatment of the neurological features of Sturge-Weber syndrome. The neurologist. 2011 Jul;17(4):179–84.
4. Raches D, Hiscock M, Chapieski L. Behavioral and academic problems in children with Sturge-Weber syndrome: Differences between children with and without seizures. Epilepsy Behav. 2012 Nov 1;25(3):457–63.
5. Schramm J, Kuczaty S, Sassen R, Elger CE, Lehe M von. Pediatric functional hemispherectomy: outcome in 92 patients. Acta Neurochir (Wien). 2012 Nov;154(11):2017–28.
6. Lo W, Marchuk DA, Ball KL, Juhász C, Jordan LC, Ewen JB, et al. Updates and future horizons on the understanding, diagnosis, and treatment of Sturge-Weber syndrome brain involvement. Dev Med Child Neurol. 2011/12/23 ed. 2012 Mar;54(3):214–23.
7. Khandelwal S, Sharma S, Patil S, Gupta D. Sturge-Weber Syndrome: A Case Report. J Dent Appl. 2014;1(5):88–90.
8. Shaikh SM, Goswami M, Singh S, Singh D. Sturge-Weber syndrome - A case report. J Oral Biol Craniofacial Res. 2015/02/19 ed. 2015;5(1):53–6.
9. Janani R, Kiruthika S, Arunachalam R, Kumaresan A. Sturge-Weber Syndrome: A Case Report. Asian J Pharm Clin Res. 2018;11(3):3–4.