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ARTIKEL LAPORAN KASUS

REHABILITATION MANAGEMENT OF A CHILD WITH AN ATYPICAL CASE OF STURGE-WEBER SYNDROME

PENATALAKSANAAN REHABILITASI ANAK DENGAN KASUS ATIPIKAL SINDROM STURGE-WEBER

Selly Christina Anggoro

Physical Medicine and Rehabilitation Department, Persahabatan General Hospital, Jl. Persahabatan Raya, Pulo Gadung, Jakarta 13230
Korespondensi: sellywijayasurya@yahoo.com

ABSTRACT
Introduction: *Sturge-Weber syndrome (SWS) is a malformation of the capillary and veins affecting the brain, the eye, and the adjacent trigeminal dermatomes of the skin, a facial port wine stain, or nevus. Type 3 SWS is a form of disease that comes without the physical facial angioma that made the disease undiagnosed and untreated optimally.*
Case Report: *We report a case of a 6-year-old girl with a history of seizures without fever since she was 13 months old. The seizure was repeated several times within a month and a year afterward but relatively controlled with routine oral medications; the last seizure attack was when she was 4 years 8 months old. Unluckily, by 6 and 3 months old, she was brought to the emergency department due to sudden focal seizure and weakness in her right extremities. The enhanced MSCT angio and brain MRI examination revealed the abnormality of the cerebral angiography and raised the diagnosis of Sturge-Weber syndrome. After several medical treatment and rehabilitation programs, her gross motor ability and balance improved with some dysfunction in fine motor ability on her right hand and few difficulties with her introductory speech. Her cognitive skill was also disturbed since she seemed to have a slower memory recall and speed processing after the latest seizure attack.*
Discussion: *As classified by Roach, et al., the clinical form presented by our patient corresponded to type 3 with exclusively neurological signs. She was previously diagnosed with epilepsy when she was 13 months old and without cutaneous angioma or ocular abnormality. The diagnosis made was based on clinical signs and supporting examinations. The management was focused on controlling symptoms and treating the disabilities caused by the disease.*
Conclusion: *SWS is a rare syndrome that needs comprehensive management, physical medicine, and rehabilitation intervention to control the symptoms and improve the patient's functional capacities.*
Key Words: *epilepsy, leptomeningeal angioma, port-wine birthmark, stroke-like episodes, Sturge–Weber syndrome.*

ABSTRAK
Pendahuluan: *Sturge-Weber syndrome (SWS) adalah malformasi kapiler dan vena yang memengaruhi otak, mata, dan dermatom trigeminal yang berdekatan pada kulit atau noda port-wine wajah atau nevus. SWS tipe 3 adalah bentuk penyakit yang datang tanpa angioma wajah fisik yang membuat penyakit ini tidak terdiagnosis dan tidak diobati secara optimal.*
Laporan Kasus: *Kami melaporkan kasus seorang gadis 6 tahun dengan riwayat kejang tanpa demam sejak dia berusia 13 bulan. Kejang berulang beberapa kali dalam satu bulan dan satu tahun setelahnya, tetapi relatif terkontrol dengan obat-obatan oral rutin, dengan serangan kejang terakhir adalah ketika dia berusia 4 tahun 8 bulan. Sayangnya, pada usia 6 dan 3 bulan, dia dibawa ke unit gawat darurat karena kejang fokal mendadak dan kelemahan pada ekstremitas kanannya. Pemeriksaan MSCT angio dan MRI otak yang ditingkatkan mengungkapkan kelainan angiografi serebral, dan meningkatkan diagnosis sindrom Sturge-Weber. Setelah beberapa kali perawatan medis dan program rehabilitasi, kemampuan motorik kasar dan keseimbangannya semakin meningkat dengan beberapa disfungsi pada kemampuan motorik halus di tangan kanannya dan sedikit kesulitan dalam berbicara dasar. Ada juga beberapa gangguan pada keterampilan kognitifnya karena dia tampaknya memiliki ingatan yang lebih lambat dan pemrosesan yang cepat setelah serangan kejang terakhir.*
Diskusi: *Seperti yang diklasifikasikan oleh Roach, et al., bentuk klinis yang disajikan oleh pasien kami sesuai dengan tipe 3 dengan tanda-tanda neurologis eksklusif yang sebelumnya didiagnosis dengan epilepsi sejak*

dia berusia 13 bulan dan tanpa angioma kulit atau kelainan okular. Diagnosis ditegakkan berdasarkan gejala klinis dan pemeriksaan penunjang. Penatalaksanaan difokuskan pada pengendalian gejala dan pengobatan kecacatan akibat penyakit tersebut.
Simpulan: *SWS merupakan sindrom langka yang memerlukan penanganan komprehensif serta intervensi medis dan rehabilitasi untuk mengendalikan gejala dan meningkatkan kapasitas fungsional pasien.*
Kata Kunci: *epilepsi, leptomeningeal angioma, tanda lahir port-wine, stroke-like episodes, Sturge-Weber syndrome*

INTRODUCTION

Sturge–Weber syndrome is a rare, sporadic neurocutaneous syndrome involving the skin, brain, and eyes, characterized by a classical triad of facial port-wine birthmark (PWB), the area innervated by the first sensory branch of the trigeminal nerve, ipsilateral leptomeningeal angiomatosis (LAM), and ocular involvement in the form of glaucoma.[1] SWS can be considered a spectrum of disease in which individuals may have abnormalities affecting all three of these systems (i.e., brain, skin, and eyes), or only two, or only one. Consequently, the specific symptoms and severity of the disorder can vary dramatically from one person to another. Symptoms are usually present at birth (congenital), yet the disorder is not inherited and does not run in families. Some symptoms may not develop until adulthood. SWS is caused by a somatic mutation, most commonly in the *GNAQ* gene. This mutation occurs randomly (sporadically) for no known reason.[2]

Due to its unique physical appearance, this illness is usually diagnosed early, during the first years of life, even though its incidence is limited. The incidence of SWS is 1:20,000 - 1:50,000 infants with no known sex or racial predilection, although it is more often underreported.[3] SWS is classified into three different subtypes by Roach, *et al*. In type I (classic, complete form), the individual demonstrates neuro-oculo-cutaneous involvement (facial angioma, leptomeningeal angioma, and glaucoma); type II SWS (bi-symptomatic form), there will be facial angioma and ophthalmic involvement without leptomeningeal angioma; and type III SWS (rarest variant, rough form), which only leptomeningeal or pial angioma present without facial or ophthalmic involvement. The incidence of type III SWS is unknown, and only a few cases are reported.[4]

A variable marks SWS but the usually progressive course in early childhood is characterized by seizures, stroke-like episodes, headaches, neurological and cognitive deterioration, hemiparesis, glaucoma, and visual field defects. More recently, the increased prevalence of otolaryngological, endocrine, and emotional-behavioral issues has been established. Neurophysiology and neuroimaging studies provide information regarding the evolution of changes in SWS over time. Early recognition and aggressive management of symptoms remain the cornerstone in managing this syndrome.[5] The mainstay of neurologic treatment is using anticonvulsants to reduce seizures. Epilepsy in SWS itself can be challenging to control, occurring in clusters of seizures and episodes of status epilepticus. Although generalized seizures are seen, most seizures in patients with SWS are focal motor with or without impaired consciousness.[6] However, in SWS without facial nevus, the

onset is usually variable and marked by manifestation of other clinical features of the classic SWS, including epilepsy in 75-90% of patients, mental retardation in 50% of patients or hemiplegia or hemiatrophy in 30% of patients.[7] Glaucoma is present in almost 30% of classical SWS. Still, in SWS without facial nevus, glaucoma may be absent because the nevus is not present in the V1 distribution. Recurrent headaches and migraine-like attacks are other common manifestations in SWS patients, reported in 37% of patients with, SWS and were variably associated with seizures. They can interfere with daily activities in a quarter of the children, and 39% of patients reported more than 1 headache attack every month. The headache was reported to be associated with glaucoma and aspirin administration, while children with monoplegia and hemiplegia were less likely to have a headache.[8]

Considering its limited and underreported incidence, the type 3 SWS would be left undiagnosed yet untreated optimally. As a consequence of being untreated, the condition could develop into severe refractory seizures, ischemic strokes, visual loss, and early cognitive impairment, which may harm and increase the risk of disabilities in the future. Therefore, a challenging multidisciplinary interaction is required for its management.

CASE REPORT

A 6 years old girl presented to the emergency unit of Persahabatan Central General Hospital with a recurrent focal seizure that lasted 5 minutes, along with weakness on the right side of her body. (Figure 1) There was a history of complaints of aching on her neck a couple days before the attack. The girl previously had medical diagnosis of epilepsy since the age of 13 months old, found out by the abnormal Electroencephalogram (EEG) which figured some cortical dysfunction in her right temporocentral and left fronto-centro-parieto-temporo-occipital regions, and epileptogenic focus on her left frontal region. The seizure was presented almost every month afterwards, until it was finally controlled with Phenobarbital and Carbamazepine by the age of 5 when her EEG was within normal limit. The history of previous trauma and bleeding diasthesis had been ruled out. She was then admitted to the pediatric intensive care unit (PICU) due to her status epilepticus during hospitalization. Her physical examination revealed marked dysarthria and right sided hemiparesis. (Figure 2) The laboratory findings showed elevation of D-dimer level (1580 ng/mL) and no signs of infection process. The enhanced-multislice computerized tomography (MSCT) of angiocerebral with contrast and the magnetic resonance imaging (MRI) of the brain revealed non-visualization superficial cortical veins with dilated deep venous drainage in her left parietal lobe with focal loss of volume in the cortical left parietal region, which on contrast provides a prominent leptomeningeal contrast enhancement, which is consistent with a Sturge-Weber Syndrome. Having ruled out differential diagnosis, the diagnosis of SWS was made based on clinical symptoms and imaging. Treatment was started using the combination of anti-epileptic

drugs phenytoin, phenobarbital, and valproic acid. She was also referred to the Physical Medicine and Rehabilitation (PM&R) department during inpatient care and received physical therapy sessions every two days. After being hospitalized for ten days, she was finally discharged home because her condition was considered stable enough.

Figure 1. Last seizure attack at 6-years-and-3-months-old

Figure 2. During inpatient care - patient was able to ambulate with hemiplegic gait and needs help to control her balance

After discharged, there was no history of epilepsy attack afterwards and the patient is still consuming her epilepsy medications until now. After 7 days discharged home, the patient was referred to the outpatient PM&R clinic. At her first visit in PM&R outpatient care, she was assessed to have some muscle weakness on her right side of the body which was considered as functional muscle strength (assessed with manual muscle testing (MMT) grade 3 for her right upper extremity and MMT grade 4- for her lower extremity). She was able to walk with the help of her mother to make her balance steady. Her Modified Wee-FIM (Functional Independence Measure) scale was considered moderate dysfunction, with difficulty to maintain walking and taking up and down the stairs due to her hemiparetic extremities. For fine motor function, she still had inadequate hand and eye coordination and three-jaw-chuck prehension that time; sparing her ability to reach, grasp, and release objects. For communication ability, she was able to communicate functionally and to perform two way interactions verbally, even though her articulation for phoneme /r/ was still not optimal yet. Her mother said that she could speak better now compared to when she was being inpatient, but her communication ability now was still not as good as it was before the attack, and her speech was still considered as slow and low in volume. The endurance

assessment result was inadequate, tested using Sit to stand test, and the result was 9 times in 30 seconds.

Apart from her medical and functional dysfunctions, we also assessed her by using the International Classification of Functioning, Disability and Health for Children and Youth, known more commonly as ICF-CY. ICF-CY is part of the WHO Family of International Classifications (WHO-FIC), play role as a standard language for children's health, education and social services. It is used to record the characteristics of child development, the environmental factors affecting child development, and developmental delays. The ICF model, as illustrated in Figure 3, integrates the major models of disability, which is multi-dimensional and interact one with another. It also recognises the role of environmental factors in the creation of disability, as well as the relevance of associated health conditions and their effects.[9-10]

The ICF perspectives from the domains of body structures, body functions, activities and participation and environmental factors from this patient can be seen in Table 1.

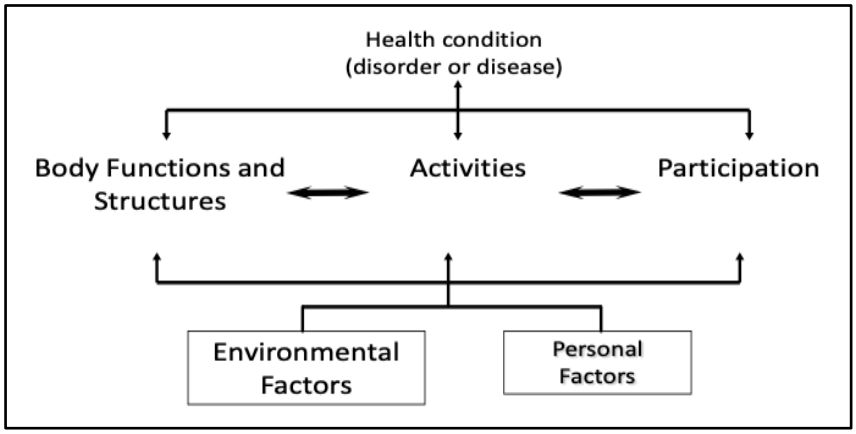

Figure 3. Interactions between the Components of ICF (WHO 2001:18)

We then gave her a routine outpatient rehabilitation therapy program which included some series of physical therapy, occupational therapy, and speech therapy which was actually should be done 2 or 3 times per week. But, unluckily, as we were still struggling with third wave of the COVID-19 Pandemic, and as a part of our infection-control programs in our hospital, we reduce the therapy frequency to be only once a week, and focusing more on educating her mother as the extension of our hand in delivering the therapy program. The physical program was focusing on the strengthening exercise for the hemiparetic extremities and muscle endurance exercise for doing ambulation activities such as standing and walking. The rehabilitation programs for the occupational therapy was to maintain good balance, both static and dynamic balance, and also for her fine motor function, to improve her wrist and hand stability, dexterity, and hand-eye coordination, especially while doing hand manipulation activities with both hands. For her speech, we

focused on giving programs to improve phoneme articulation and increase her expressive language function to obtain functional communication skills.

Table 1. ICF-CY Perspectives of Atypical case of Sturge Weber Syndrome

ICF - Body Structures	ICF - Body Functions	ICF - Activities & Participation	ICF - Environmental Factors
s110 Structure of brain s198 Structure of the nervous system, other specified s230 Structures around eye s799 Structures related to movement, unspecified	b117 Intellectual functions b140 Attention functions b164 Higher-level cognitive functions b210 Seeing functions b320 Articulation functions b455 Exercise tolerance functions b499 Functions of the cardiovascular, haematological, immunological and respiratory systems, unspecified b730 Muscle power functions b740 Muscle endurance functions b789 Movement functions, other specified and unspecified b799 Neuromusculoskeletal and movement-related functions, unspecified	d137 Acquiring concepts d175 Solving problems d177 Making decisions d240 Handling stress and other psychological demands d398 Communication, other specified d460 Moving around in different locations d540 Dressing d720 Complex interpersonal interactions d820 School education d830 Higher education d835 School life and related activities d880 Engagement in play	e320 Friends e325 Acquaintances, peers colleagues, neighbours and community members e420 Individual attitudes of friends e425 Individual attitudes of acquaintances, peers colleagues, neighbours and community members

Figure 4. (a) At 3 months post inpatient care - patient able to use fine motor skills adequately with three-jaw chuck pattern

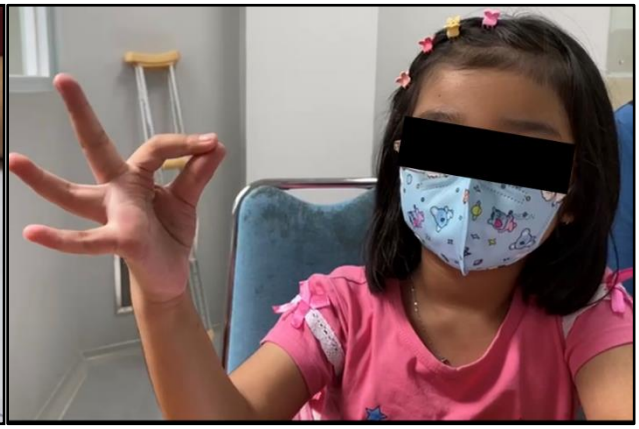

Figure 4. (b) Her fine motor coordination was getting better now

During monthly evaluation, she had progressed aggressively and presented rapid motor recovery after her first month's sessions of therapy. She was able to walk more steady and unaided anymore. Her fine motor skills was also improving, as her hand and eye coordination being improved, with only little weakness on the intrinsic muscles of her right hand. (Figure 4) She was able to understand and comprehend our conversation well during our consultation session, and her cognitive function was considered to have good enough comprehension for simple receptive language abilities, and she was also able to response

with proper verbal expressive language, although her voice seemed low in volume and quite slow and flat in tone. For this reason, her speech and occupational therapy sessions would be continued in order to improve her dexterity skills and voice quality.

From further anamnesis, the patient's mother said that there was a change in patient's cognitive ability. Before sick, the patient was able to interact well and follow instructions. She was able to recognize letters, numbers, and read words in sentences easily. But since the latest seizure attack and stroke-like syndrome, the patient was unable to respond to others' with her usual speed. We finally sent the patient to a psychologist to get a thorough psychological examination and a school preparation test knowing that she was actually prepared by her parents to enter the elementary school this year. From the psychological assessment, it was revealed that her intelligence level was inadequate, with her general intelligence quotient was 80 (under average, slow learner), her verbal and theoretical intelligence was 48 (low, moderate mental retardation), and her performance or practical intelligence was 92 (average). She had good enough emotional maturity, inadequate fine motor and social maturity but good ability to follow authority. She was also suggested to suspend the entrance to the elementary school by this year and explore her ability to be involved in daily programs more before going to elementary school by the psychologist. Unluckily, we do not have the objective data measurement for cognitive function before the disease, and could not compare before and after sick. But, as a conclusion, this cognitive dysfunction was consistent with the possibility of cognitive dysfunction found in SWS patients in many parts of the world

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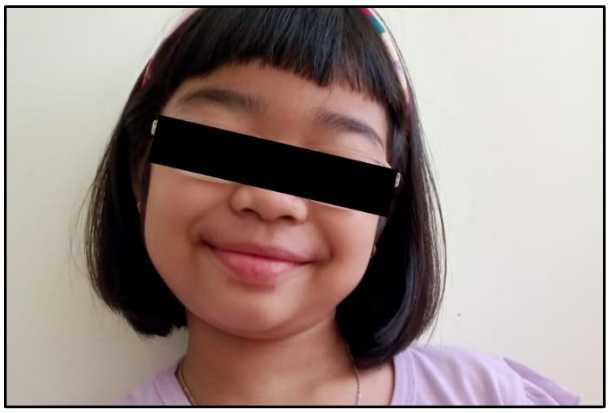

Figure 5. Note her normal face, without the port-wine birthmark which is usually seen in Sturge-Weber syndrome

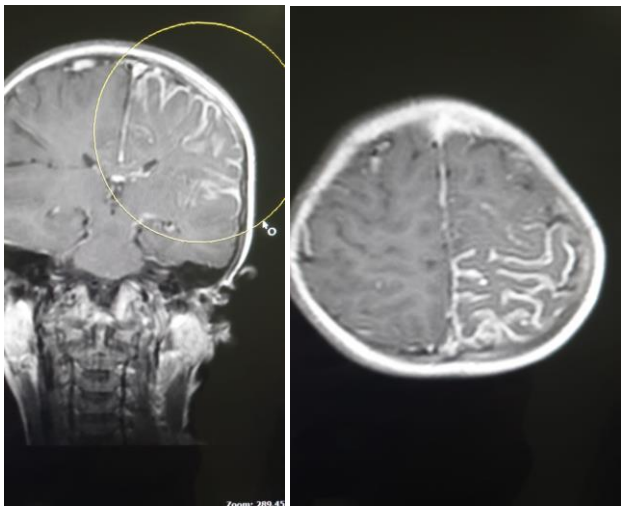

Figure 7. Focal loss of left parietal cortex volume that shows the prominent leptomeningeal contrast enhancement (pial angiomatosis) and non-visualized superficial cortical veins with dilatation of deep venous drainage at left parietal region that supports the appearance of Sturge-Weber syndrome.

DISCUSSION

The diagnosis of SWS brain involvement requires a contrast-enhanced MRI, and susceptibility-weighted imaging and post-contrast flair sequences may increase the sensitivity. The brain involvement of SWS is made by visualizing the enhancing leptomeningeal vessels as it was seen in this case, although the typical facial PWB is absent. Strokes and stroke-like episodes which are common, particularly in infants and toddlers, can be defined as a focal neurologic deficit lasting longer than 24 hours; these impairments resolve over a period of days to weeks. Neurologic impairment that does not fully resolve but persists may be referred to as a stroke. Many children gradually acquire a hemiparesis in a stepwise fashion such that stroke events are not recognized; other children have clear acute onset of focal neurologic deficit. During a stroke-like episode the EEG often demonstrates focal slowing. Treatment with anticonvulsant or low-dose aspirin or both before the onset of seizures is an option, with surgical resection may be offered to those whose seizures are medically refractory. In this case, the medical treatment was quite successful as the seizure was better controlled with combination of anti-seizure agents. Other things that should be managed are the endocrine, medical rehabilitation and cognitive comorbidities.[6]

From the 3 types classified by Roach *et al*., the clinical form presented by our patient corresponded to type 3 with exclusively neurological signs without cutaneous angioma or ocular abnormality.[4] In most SWS cases without facial angioma, epilepsy is the presenting symptom with leptomeningeal angiomatosis and calcification evident on initial imaging[10], which was also consistent with our patient that was previously diagnosed with epilepsy since she was 13 months old of age. It suits with the literature that said seventy five to 90% of children with SWS have focal seizures often misdiagnosed as an Epilepsy, that are initially observed in most children who have SWS, most of which will present as a transient episodes of hemiparesis.[11]

Physical medicine and rehabilitation (PM&R) is the medical specialty concerned with restoring and maintaining the highest possible level of function, independence, and quality of life.[13] Also known as physiatry or rehabilitation medicine, it aims to enhance and restore functional ability and quality of life to those with physical impairments or disabilities affecting the brain, spinal cord, nerves, bones, joints, ligaments, muscles, and tendons. Unlike other medical specialties that focus on a medical “cure,” the goals of the physiatrist are to maximize patients’ independence in activities of daily living and improve quality of life.[14] In this case, after series of rehabilitation programs during inpatient and outpatient care, the rapid functional progression was finally gained and the patient was able to mobilize and perform activity daily living independently with minimal assistance. As a part of disease management, the treatment goals in this syndrome are to minimize or eliminate seizures and to maximize intellectual potential[15], due to the literature saying that approximately half these

children have frank mental retardation, whereas others display learning disabilities, attention disorders, or behavioral disturbances.[11, 16] That is why symptoms control along with the assessment and intervention for cognitive function are considered very important to be noted in this syndrome, as it was also noted in case vignettes that highlight the seizure control, return to functional baseline and subsequent further skill development.[16]

CONCLUSION

As a part of seizure control and to improve the functional disabilities after stroke-like attack, the medical treatment along with the rehabilitation programs is routinely adviced as an intervention treatment control for Sturge-Weber Syndrome.

Although the patient's cognitive dysfunction itself was not optimally resolved yet right now, we hope that better progression will be enhanced with the upcoming routine cognitive rehabilitation programs along with the psychological consultation sessions. We also hope that in the future, new therapeutic options are likely to be offered stemming from preclinical studies and small pilot clinical trials currently ongoing.

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