Sturge Weber Syndrome: Abnormalities in the Brain, Skin and Eyes from Birth - A Case Report

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Abstract: A 23-year-old woman was referred from neurologic outpatient clinic with Hemiparesis Dextra (atrophy) e.c Sturge Weber Syndrome. She complained weakness of right extremities since she was a child (4 month-old) after she experienced a seizure. She felt easily tired after walking 100 meter, especially on the right leg. There was numbness on right face and extremity. She felt pain on right eye, and routinely controlled to Ophthalmology department. There was red skin on the left face. Physical examination revealed numbness, weakness, atrophy, increased physiological reflex and spasticity on right extremities. There was also numbness on her right face. Redness on the left face (port wine stain) and eye (glaucoma and buftalmos), pain on left eye and blurred vision. There was parese on left facial, vestibulocochlear, glossopharynx and hipoglossus nerve. MRI Brain discovers Sturge Weber Syndrome. The rehabilitation program consists of ROM exercise, strengthening exercise, sensory resensitisation, hand function exercise, breathing exercise, balance exercise and gait training.

1 INTRODUCTION

Sturge-Weber Syndrome is a sporadic, congenital neurocutaneous disorder, characterized by intracranial leptomeningeal vascular malformations associated with a facial port wine stain (nevus flaemmus) (Chhabria et al, 2017). Sturge–Weber syndrome (SWS) or encephalotrigeminal angiomatosis is a congenital, non-hereditary, condition of unknown etiology. The disease shows facial port-wine stain, ocular abnormalities (glaucoma and choroidal hemangioma), and leptomeningeal angioma. It belongs to a group of the disorder known as the phacomatoses ("mother-spot" diseases). SWS was first described by Schirmer in 1860 and later more specifically by Sturge in 1879. He associated dermatological and ophthalmic changes of the disease to neurologic symptoms. Weber, in 1929, stated the radiologic alterations seen in patients of SWS. (Kulkarni, 2015).

It is rare disorder occurring with no racial predilection equally affecting males and females the leptomeninges epilepsy, Port-wine stain, ocular involvement, dermal angiomas, mental retardation, hemiplegia, and abnormalities in skull radiographs (Kulkarni, 2015).

The syndrome occurs almost always sporadically and have no definite hereditary influence. Patient with unilateral facial port wine stains of upper eye lid increases the risk of glaucoma and regularly had seizures and hemiparesis of the contralateral side and that is the result of an intracranial hemangioma. Sturge weber syndrome appear as a congenital lesion usually benign tumors of blood vessels and have no chance of malignant transformation (Moly et al, 2016).

2 CASE PRESENTATION

A 24 years old woman, Javanese from Lamongan city, East Java. She hasn't married yet with low economic stage and doesn't work. She was referred from neurologic outpatient clinic with weakness of right extremities (atrophy) because of Sturge Weber Syndrome. She complained weakness of right extremities since she was a child (4 month-old) after she experienced a seizure (figure 1C). She felt easily tired after walking 100 meter, especially on the right leg. There was numbness on right face and extremity. She felt pain on right eye, and routinely controlled to Ophthalmology department. There was

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red skin on the left face. There was no abnormality of prenatal and natal history. Post natal history, there was red skin on left face, asymmetric face and left eye is more prominent (figure 1A&B). Her development was similar to another child. She experienced seizure routinely since 1 month ago, twice a day. She often felt pain on her left head in the last 3 years ago. In 2002, she underwent glaucoma surgery.

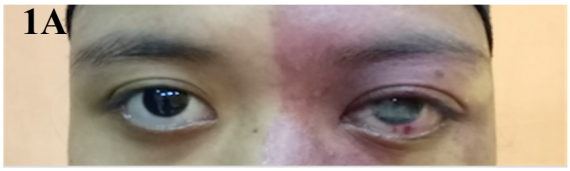

Figure (1A): Port-wine stain, glaucoma and buftalmos.

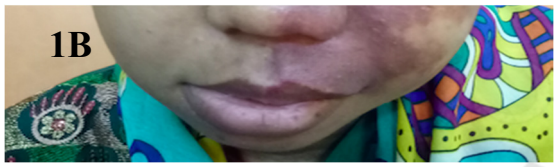

Figure (1B): Port-wine stain, orbicularis oris weakness

Figure (1C) : Atrophy right arm and leg

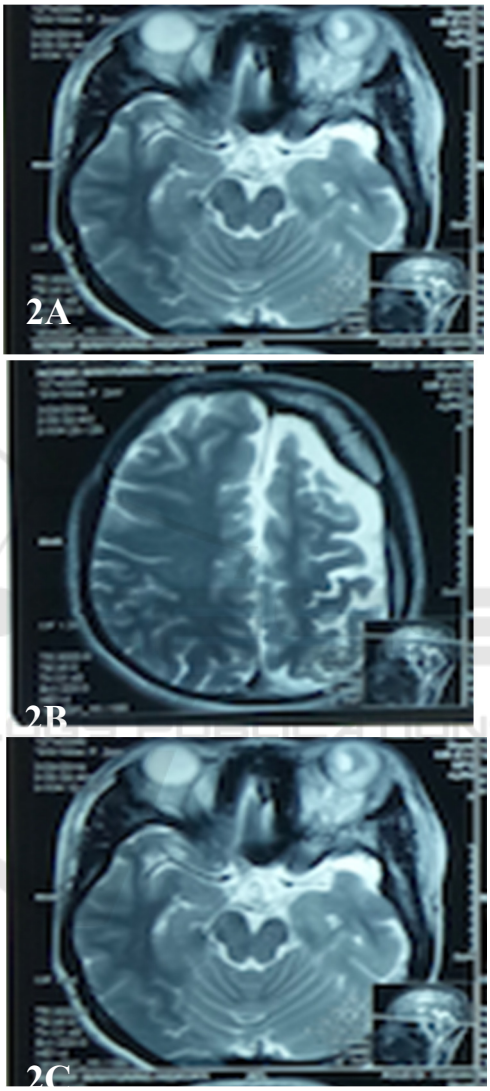

Figure (2A, B, C) : MRI brain : Sturge Weber Syndrome

On examination, we found numbness, weakness, and atrophy of right extremities with Full range of movement (ROM) and the manual muscle test (MMT) of right wrist flexor was 1, fingers extensor was 2, thumb flexor and extensor were 1. The other still functional. There was increased physiological reflex and spasticity on right extremities with Modified Ashworth Scale (MAS) were 1+ on elbow flexor and extensor, MAS 1 on wrist flexor and extensor, MAS 2 on knee extensor. There was also numbness on her right face. Redness on the left face (port wine stain) and eye (glaucoma and bultalmos), pain on right eye and blurred vision. There was parese of left facial, vestibulocochlear, glossopharynx and hipoglossus nerve. She can't do tandem walking. Her dynamic standing balance also

was poor. Hand function like spherical, cylindrical, lateral tip and pinch were non functional. Count test was 23 and chest expansion was still good. MRI brain with Sturge Weber Syndrome (figure 2A, B, C).

We give rehabilitation program that consist of ROM exercise, strengthening exercise, sensory resensitisation, hand function exercise (especially for activity daily living), breathing exercise, balance exercise and gait training and posture correction.

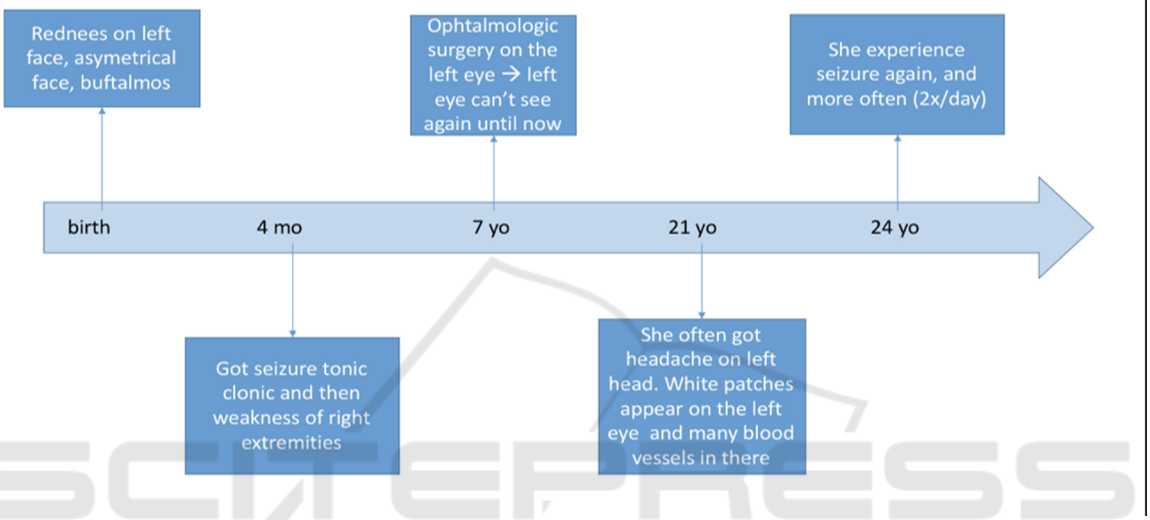

Figure 3: Course of the disease.

We suggest this intervention two times per week (in Lamongan hospital) and evaluated in Soetomo Hospital every month. We educated the patient to do exercise at home every day.

In the first month of rehabilitation program, our main goal was to improve the posture, maintenance the ROM, improve the gait dan hand function. Due to the socioeconomic condition and her house far from hospital, she was given a home-based exercise program and therapy to the hospital in every week. The program would be evaluated every 1 month.

Two months later, the patient was evaluated in Hospital, there was no significant increasing the MMT, but the patient felt more comfort in walking. For balance test, we tried to give one leg stand, and she can do this just on the left leg. The right leg still weak to support whole her body. We also evaluated hand function, but there was no significant increase. She said that she did the exercise program was not optimal because she still focused on eye treatment.

The right eye very painful and she routinely control to ophthalmology department.

3 DISCUSSION

Sturge-Weber syndrome (SWS) is a congenital, sporadically occurring, neurocutaneous syndrome that presents classically with port-wine stain, leptomeningeal angiomas, and glaucoma. The systemic implications of SWS are vast and involve not only ophthalmic manifestations but also dermatologic, neurologic, and oral manifestations. Neuroimaging, in particular, plays an important role in the diagnosis and management of this disease (Maslin, 2014). An estimated frequency of 1 per 50,000 live births have SWS, although experts believe many more people have the disorder but have not yet been identified. The hallmark intracranial vascular anomaly is leptomeningeal angiomatosis, most often involving the occipital and posterior parietal lobes, but it can affect other cortical regions and both cerebral hemispheres. An ipsilateral facial cutaneous vascular malformation usually affects the upper face in a distribution consistent with the ophthalmic division of the trigeminal nerve. Other clinical findings associated with SWS are seizures, glaucoma, headache,

transient stroke like neurological deficits, and behavioral problems. Hemiparesis, hemiatrophy, and hemianopia may occur contralateral to the cortical abnormality.

Children with SWS suffer from a variety of neurologic abnormalities, including epilepsy, mental retardation, and attention-deficit hyperactivity disorder, migraine, and stroke like episodes. Seventy five to 90% of children with SWS have epilepsy. Focal seizures are initially observed in most children who have SWS. Fever and infection often precipitate seizure onset. If noncontrasted computed tomography obtained in the emergency room setting after seizure activity is reported as normal or reveals focal calcification ipsilateral to a cutaneous angioma, more complete cerebral imaging is warranted. Most seizures are focal, because the lesion responsible for the epilepsy in SWS is focal. Seizures are likely caused by hypoxia and microcirculatory stasis. Children with radiographic findings of intracranial angiomatosis usually develop seizures by the age of 3 years. Approximately half these children have frank mental retardation, whereas others display learning disabilities, attention disorders, or behavioral disturbances.

SWS classically presents with a unilateral cutaneous nevus PWS or red wine stains on the face. The cutaneous presentation occurs due to early embryonic vascular malformation. SWS may also present with angiomas in the leptomeninges resulting in epilepsy and hemiparesis and/or angiomas in the eye causing glaucoma. The most frequent oral presentation of SWS is hyperplasia of the gingiva, affecting the maxilla, floor of the mouth, lips, cheeks, palate and tongue of the same side. SWS may also present with changes in the histology and morphology of gingiva, periodontium, and pulp. (Neerupakam, 2017)

It's typical manifestations include, cutaneous-port wine stain on the face, ocular-glaucoma, choroidal hemangioma and neural features-leptomeningeal haemangioma, seizures. Oral involvement in SWS presents as a gingival haemangiomatous lesion limited to the maxilla and mandible of the same side. (Neerupakam, 2017)

SWS can be classified into three different types:

- Type 1 (most common type) is characterized by port-wine stain, cerebral malformation (leptomeningeal angiomas), and the possibility of glaucoma or choroidal lesions. Seizures may occur during the first year of life. Developmental disabilities may be seen during the first year.
- Type 2 is characterized by port-wine stain and possibly glaucoma without cerebral malformation (leptomeningeal angiomas). Headaches or migraines may also occur.
- Type 3 is characterized by cerebral malformation (leptomeningeal angiomas) exclusively. Port-wine stain is not present and glaucoma is rare. (Hernandez, 2019)

In this case we got the patient has weakness of right extremities, unable to use right hand for normal activity, pain on right eye, redness on left eye (glaucoma and buftalmos), red skin on left face (port-wine stain), blurred vision on left eye. Weakness on left facial nerve, vestibulocochlear nerve, glossopharynx nerve and hipoglossus nerve. MRI brain support to Sturge Weber Syndrome. We give rehabilitation program to improve her quality of life. The patient felt more comfort to walking after 2 months, and hand function is still not improving. There was no adverse events or harm while running this rehabilitation program.

She runs her rehabilitation program at a hospital near her home, and every months control to Soetomo Hospital. She didn't do this rehabilitation program maximally because she focused on her eye treatment first that painful and very disturb her. Although there was no significant increase in MMT, but she felt more comfort in walking and not easily tired as before.

The prognosis in SWS varies widely. Although patients with widespread hemispheral disease or bihemispheric disease are at greatest risk for neurologic complications, many function virtually normally. Clearly, a subgroup of patients with limited central nervous system involvement as defined by neuroimaging studies has a particularly malignant clinical course, with intractable epilepsy, headache, stroke like episodes, and cognitive deterioration. There is a greater likelihood of intellectual impairment when seizure start before the age of 2 and are resistant to treatment. Prognosis is worse in the minority of children who have both side of the brain affected by the blood vessel abnormality (NINDS, 2019).

4 CONCLUSIONS

It is important to know about the disease to know the treatment and the obstacle for our rehabilitation program. We give this program to the patient to help her improve the quality of life. Although the course of the disease will go on and cannot be cured. We

just help patient to get comfort in her daily activity and adaptation for her condition.

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