

Sturge-Weber Syndrome Coexisting With Moyamoya Disease in the Fifth Decade

A Case Report and Literature Review

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Abstract: Sturge-Weber syndrome (SWS) is a sporadically occurring neurocutaneous syndrome characterized by port-wine stain over the face, ocular abnormalities (glaucoma and choroidal hemangioma), and leptomeningeal angiomas. It is usually diagnosed in infancy, but it may occasionally present in adulthood with seizures or stroke-like episodes. Here, we report a 46-year-old male patient, having SWS coexisting with moyamoya disease, attending our hospital due to sudden loss of consciousness. We also searched PubMed (from its earliest date to August 2014) for case reports mentioning that SWS presents in adulthood. We identified 31 patients. The common clinical manifestation are seizures, stroke-like episodes, and migraine-like headaches. On the basis of our findings in this patient, we would recommend that patients with a port-wine nevus of the face should be given further investigation to rule out an intracranial vascular malformation, especially if seizures, stroke-like episodes, or migraine-like headaches are present.

Key Words: Sturge-Weber syndrome, moyamoya, adults, case report
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Sturge-Weber syndrome (SWS) is a sporadically occurring neurocutaneous syndrome characterized by port-wine stain over the face, ocular abnormalities (glaucoma and choroidal hemangioma), and leptomeningeal angiomas with a frequency of ~1 per 50,000.^{1–3} The manifestation of SWS is often partial or incomplete. Therefore, SWS is divided into 3 types: (1) type I—with facial, choroidal, and leptomeningeal angioma and possible glaucoma (classic form); (2) type II—with facial angioma without evident endocranial involvement; and (3) type III—with exclusive leptomeningeal angioma.⁴ It is usually diagnosed in infancy, but it may occasionally present in adulthood with seizures or stroke-like episodes. Here, we report a 46-year-old male patient, having SWS coexisting with moyamoya disease (MMD), attending to our hospital due to sudden loss of consciousness. Informed consent of the patient was obtained.

CASE PRESENTATION

A 46-year-old man was admitted to our hospital because of sudden loss of consciousness. Eight months ago, he was presented to

the neurosurgery department of our hospital due to headache accompanied by loss of consciousness. Computed tomography (CT) of the head showed the left basal ganglia hemorrhage rupturing into the ventricle (figure not available). After treatment, he recovered, and was discharged home. He also had a history of epilepsy presenting tonic-clonic seizure with loss of consciousness 20 years ago. Phenytoin was taken to control symptoms. Three years ago, he stopped the medicine, and had 2 similar attacks.

On examination, he was noted to have a facial angioma (Fig. 1) that was present at birth. Orientation, attention, and calculation are impaired. His mini-mental state examination score was 17. There was weakness of the right upper extremity and right hemidysthesia. Neck stiffness and Kernig's sign were positive.

Laboratory investigations including hemogram, liver and kidney functions, C-reaction protein, erythrocyte sedimentation rate, thyroid function tests, antineutrophil cytoplasmic antibodies, and human immunodeficiency virus test were normal. Cerebrospinal examination showed the appearance was red with $3780 \times 10^6/L$ RBC, cell counts were $6 \times 10^6/L$, protein was 0.50 g/L, and glucose and chlorine were normal. The ultrasound of abdomen was normal. Head image examinations are shown in



FIGURE 1. Port-wine stain over the face in the patient.

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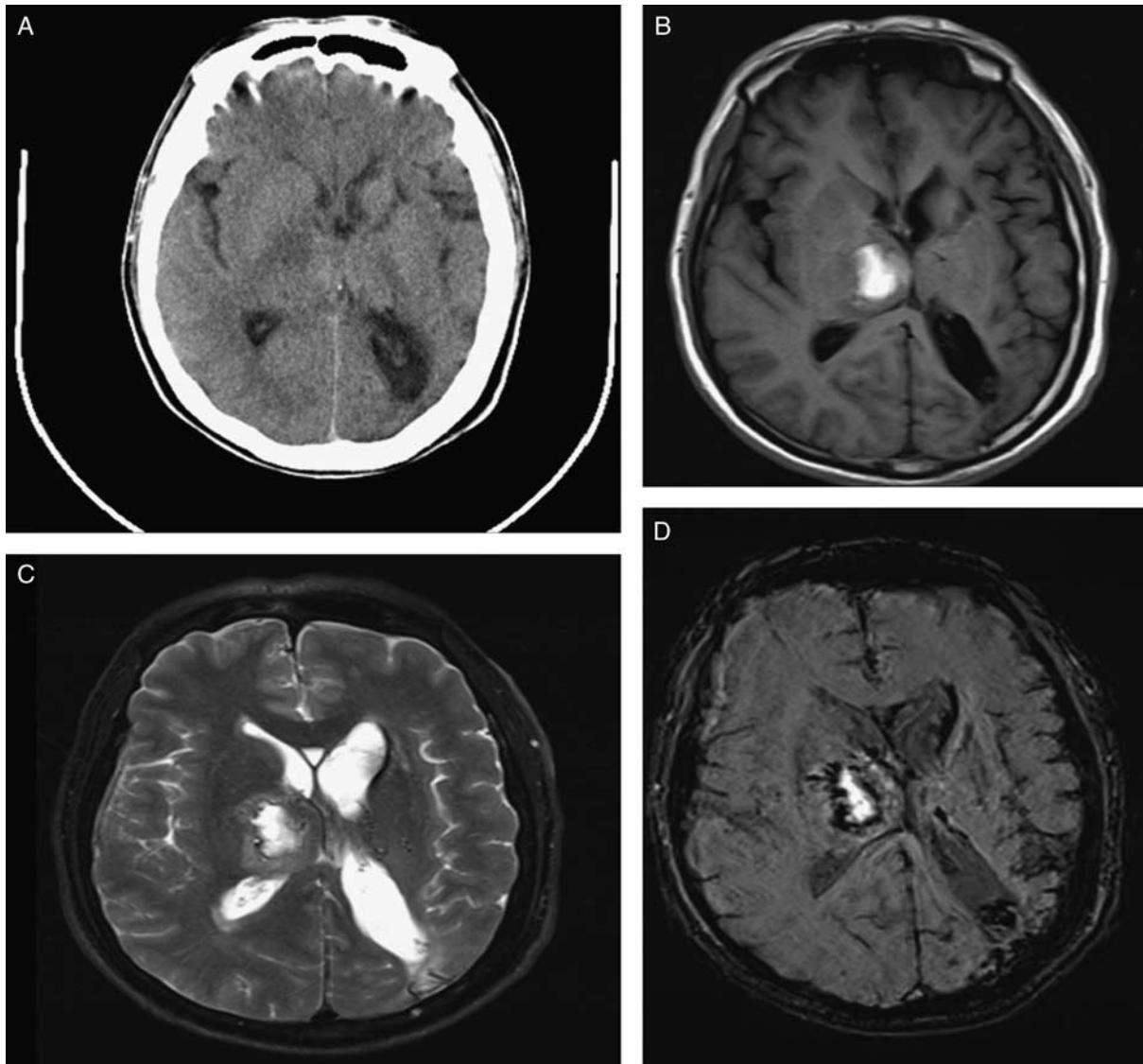


FIGURE 2. A, Axial noncontrast computed tomographic scan of the head showed bilateral lateral ventricles' asymmetry with left side expanded, and the left temporal lobe, occipital lobe atrophy. B and C, Axial noncontrast magnetic resonance scan of the head showed hemorrhage in the right thalamus and the bilateral lateral ventricles asymmetry with left side expanded. D, Axial noncontrast susceptibility-weighted imaging scan of the head showed old hemorrhage in the right thalamus and left lateral ventricles.

Figures 2A–D and 3. During hospitalization, Topiramate was given to control seizures; blood pressure and fever were controlled. He recovered and was discharged home after 1 week. At 3 months, the patient did not have loss of consciousness and seizures.

We also searched PubMed (from its earliest date to August 2014) for case reports mentioning that SWS presents in adulthood. We identified 31 patients. Age ranged from 21 to 61 years. Male sex accounted for 48.4% (15/31). The prevalence of SWS types are as follows: type I, 74.2%; type II, 3.2%; and type III, 9.7%. The common clinical manifestations are seizures, stroke-like episodes, and migraine-like headaches (Table 1).

DISCUSSION

In this report, we present a patient with SWS coexisting with MMD in adulthood. It is difficult to evaluate the relationship and their effect on each other. We did not find the underlying disease process contributing to the formation of moyamoya in this patient.

The patient first presented headache accompanied by loss of consciousness. Left basal ganglia hemorrhage was confirmed by head CT. However, he was not given further examination. He was admitted to our hospital for a second time due to sudden loss of consciousness. We considered the diagnosis of subarachnoid hemorrhage on the basis of positive of cerebrospinal examination, although head CT scan was negative. We proposed a further evaluation of vessels. Digital subtraction angiography showed bilateral internal carotid artery occlusion accompanied by abnormal vascular formations. Management of seizures and stroke-like episodes is the key for preventing progression of neurological injury in SWS. Anticonvulsants remain the cornerstone in managing seizure.³⁶ Studies on the pathophysiology of SWS have clearly highlighted the contribution of venous congestion, stasis, and thrombosis in the symptomatology of progressive brain injury in this disease.⁴ Hence, low-dose aspirin may be considered, although there is lack of evidence.

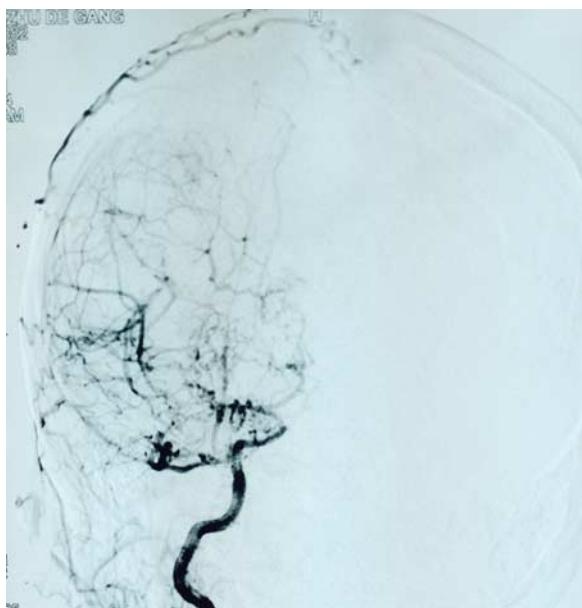


FIGURE 3. Digital subtraction angiography showed bilateral internal carotid artery occlusion accompanied by abnormal vascular formations consistent with moyamoya disease.

MMD is a progressive intracranial arteriopathy, characterized by bilateral stenosis of the distal portion of the internal carotid artery and the proximal anterior and middle cerebral arteries, resulting in transient ischemic attacks or strokes. Clinically, children with MMD present with ischemic attacks, and adults present with either ischemic or hemorrhagic events.³⁷ The pathogenesis of MMD remains unresolved, but thickened intima of the major branches of the circle of Willis, and moyamoya vessels was found. Currently, there is no treatment that can halt progression or reverse the intracranial arteriopathy of MMD. Treatment strategies are aimed at alleviating symptoms and preventing occurrence of strokes or transient ischemic attack.^{38,39} Many practitioners would like to prescribe antiplatelet agents, although it is unclear whether antiplatelet agents improve outcome. In addition, the surgical revascularization is considered as the treatment of patients with MMD.⁴⁰ Recently, a meta-analysis including 16 studies showed that surgical treatment significantly reduced the risk of stroke, which also depends on more robust evidence to bring more benefits for these patients.⁴¹ But in this patient, intracranial hemorrhage occurred twice; hence, we did not prescribe the antiplatelet agents. Supportive treatments including controls for fever, pain, and seizure medication were given.

On the basis of our findings in this patient, we would recommend that patients with a port-wine nevus of the face should be given further investigation to rule out an intracranial

TABLE 1. Summary of Reported Cases of Patients With SWS in Adulthood

References	Age (y)	Sex	Clinical Manifestation	Type
Traub ⁵	58	Female	Migraine and seizure	SWS type I
Chen ⁶	50	Male	Epilepsy	SWS type I
Ferrari ⁷	56	Male	Tonic-clonic seizure with loss of consciousness	SWS type I
Czarnecki ⁸	53	Male	The left eye was blind and focal epilepsy	SWS type I
Hussain ⁹	45	Male	Left homonymous hemianopsia	SWS type I
Choi ¹⁰	20	Female	Gingival enlargement and pain on the upper jaw	SWS type I
Gadit ¹¹	22	Male	Seizure, mental retardation	SWS type I
Utsunomiya ¹²	23	Female	Seizures and severe temper tantrum	SWS type I
Madaan ¹³	82	Male	Complex partial seizures	SWS type I
Kumar ¹⁴	22	Female	Acute left hemiparesis	SWS type I
Dora ¹⁵	22	Male	Throbbing right-sided headache	SWS type I
Castilla-Guerra ¹⁶	40	Female	Gastrointestinal hemorrhage	SWS type I
Freilinger ¹⁷	21	Male	Hemiplegic migraine	SWS type I
Bovo ¹⁸	35	Female	Seizure and hearing loss	SWS type I
Jung ¹⁹	29	Female	Left-sided weakness associated with headache	SWS type I
Jacobs ²⁰	41	Male	Migraine accompanied by visual scotoma and hemiparesis and seizure	SWS type I
Aquilia ²¹	37	Female	Hemorrhage in the left temporal lobe	SWS type I
Lisotto ²²	59	Male	Migraine-like headaches	SWS type I
Knapp ²³	22	Male	Left central retinal vein occlusion	SWS type I
Dolkart ²⁴	24	Female	Complex partial seizures	SWS type I
Zhou ²⁵	23	Male	Epilepsy	SWS type I
Neki ²⁶	23	Female	Tonic-clonic seizure	SWS type I
Martínez-Gutiérrez ²⁷	34	Male	Left ocular pain and left chronic exophthalmos	SWS type I
Niemczyk ²⁸	50	Female	Autosomal dominant polycystic kidney disease	SWS type II
Kapadia ²⁹	38	Female	Left-sided headaches	SWS type III
Kim ³⁰	46	Female	Generalized seizure with the loss of consciousness	SWS type III
Huang ³¹	36	Female	Migraine-like attacks with prolonged visual field defect	SWS type III
Alli ³²	56	Female	Pain, photophobia in the left eye	Unclear
Planche ³³	40	Female	Hemiplegic migraine-like attacks	Unclear
Matysik-Woźniak ³⁴	61	Male	Basal cell carcinoma in nevus flammeus	Unclear
Ye ³⁵	65	Male	Visual loss of the left eye	Unclear

SWS indicates Sturge-Weber syndrome.

vascular malformation, especially if seizures, stroke-like episodes, or migraine-like headaches are present.

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