12-Year Retrospective Study of Sturge-Weber Syndrome and Literature Review

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Objective: To review the clinical manifestations and neuroimaging features of patients with Sturge-Weber syndrome (SWS) treated at Srinagarind Hospital over a 12-year period.

Material and Method: A retrospective study of sixteen patients with SWS (9 males and 7 females) was conducted. The medical records, photographs, and neuroimaging studies were reviewed.

Results: All patients had port-wine stain (PWS) involving the eyelid. Bilateral cutaneous lesions were revealed in four patients (25%). Glaucoma was the main ocular disease being diagnosed in 11 eyes of nine patients (56.25%); four eyes were finally blind. The cyclodestructive procedure and/or surgical treatment was required in four eyes. Other ocular abnormalities were refractive error, dilated episcleral vessels, corneal abnormalities, tortuous retinal vessels, choroidal hemangioma, amblyopia, and strabismus. Twelve patients (75%) had neurological impairment including seizure, hemiparesis, headache, and delayed development. However, the most common neurological manifestation was epilepsy (75%), which could be controlled with antiepileptic drugs. Neurological imaging was performed in the majority of cases (14 patients). Intracranial abnormalities were demonstrated in 11 patients (78.57%). These included cerebral atrophy (81.82%), cerebral calcification (54.55%), leptomeningeal angioma (27.27%), and enlarged choroidal plexus (27.27%). The ocular complication and intracranial abnormalities were usually ipsilateral to the PWS. One patient with unilateral PWS, however, had bilateral intracranial lesion.

Conclusion: Port-wine stains, glaucoma, and seizure were the most common clinical features of Sturge-Weber syndrome detected in the present study. Complete ophthalmic and neurological evaluation should be performed at the time of diagnosis. Multidisciplinary team management as well as lifelong follow-up is needed.

Keywords: Sturge-Weber, Port-wine stains, Nevus flammeus, Leptomeningeal angiomatosis, Epilepsy

Sturge-Weber syndrome (SWS), also known as encephalotrigeminal angiomatosis, is a rare sporadic neurocutaneous syndrome. The classic triad is characterized by port-wine stain (PWS) that affects the facial area innervated by the first sensory or ophthalmic branch of the trigeminal nerve (V1), ipsilateral leptomeningeal angiomatosis that most often involving the occipital lobes and posterior parietal lobe, and ocular vascular malformation. Glaucoma is the most common ophthalmic complication of SWS, and then early diagnosis and treatment can prevent the blindness. However, the clinical manifestations of SWS are varied widely, with absent or varying neurological and/or ocular features in some cases[1]. Although SWS is among the common phakomotosis, the pathogenesis remains unclear and no true animal model exists.

Material and Method

All patients with SWS who were treated at Srinagarind Hospital between November 2001 and July 2013 were studied. The study was approved by the Khon Kaen University Ethics Committee. The medical records, photographs, and neuroimaging images of patients were reviewed. Port-wine stains were topographically analyzed and mapped to the sensory distribution of division(s) of the trigeminal nerve.

Results

Sixteen patients with SWS were identified, nine patients (56.25%) were males and seven (43.75%)

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were females. The age of the patients ranged from newborn to 35 years old. All patients had PWS on their face with neurological and/or ocular complications. Twelve patients (75%) had neurological impairment and ten patients (62.50%) had ocular problem. One patient also had extensive cutaneous capillary malformations on the right side of the body and along the right leg as well as limb hypertrophy which were consistent with Klippel-Trenaunay-Weber syndrome. One patient had multiple large Mongolian spots on his back, the abdomen, and along the right thigh and, thus, was diagnosed with Phacomatosis pigmentovascularis type IIb. Subsequent examinations and treatments were conducted during the period of six months to 20 years.

All patients had PWS affecting the first sensory branch of trigeminal nerve (V1) distribution. However, only four patients (25%) had bilateral facial cutaneous vascular malformation (Fig. 1). Unilateral PWS often located in both V1 and V2 facial areas. The ocular complication was usually ipsilateral to the PWS although the ocular examination was not done in three children. Glaucoma was the main ocular disease observed in 11 eyes of nine patients (56.25%). Although glaucoma was related to the extension of the skin lesion in the palpebral area, bilateral glaucoma was found in one patient who had unilateral PWS affecting V1 area. Age at onset of glaucoma was during the infancy in three patients (33.33%), between one and four years in two patients (22.22%) and after four years of age in four patients (44.44%). The majority of cases could be treated with anti-glaucomatous medication, however, cyclodestructive procedure was needed in three eyes and surgical treatment was performed in two eyes. During the follow-up period, frequent ophthalmological examinations with various medical and surgical treatments could not prevent an important loss of visual acuity in most patients; and four eyes were finally blind.

Refractive error was revealed in eight eyes, most of them had mild refractive error. Dilated episcleral vessels were demonstrated in seven patients (43.75%). Corneal findings associated with infantile glaucoma were demonstrated in six eyes of four patients (25%) including corneal haziness, megalocornea, and buphthalmos. Choroidal hemangioma were often developed in patient with SWS whose PWS involved all branches of trigeminal nerve (V1-3) distribution. Diffuse choroidal hemangioma, which is a characteristic feature, was found in four eyes of three patients (18.75%) and one of them also presented with bullous retinal detachment. At fundus

examination, it appeared as a diffuse or localized elevation of the dark red retina, causing blurred disc because of the difference in planar of focus (Fig. 2). The other posterior segment finding in the present study was tortuous retinal vessel.

Twelve patients (75%) had neurological impairment. However, the most common neurological manifestation detected in the present study was epilepsy (75%). Ten of twelve patients (83.33%) had their first seizure during their infancy, from two to 11 months old. The other neurological findings included hemiparesis (37.50%), delayed development, cluster headache, and congenital hearing loss. Most
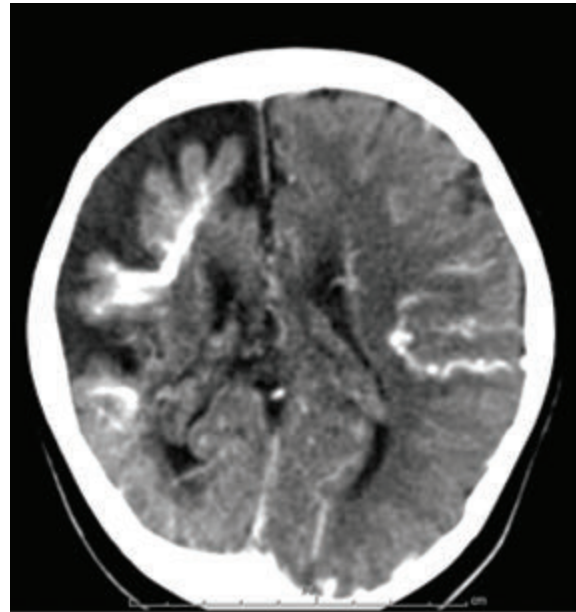

Fig. 1   Bilateral extensive port-wire stains.

Fig. 2   Diffuse choroidal hemangioma spares the superotemporal retina.
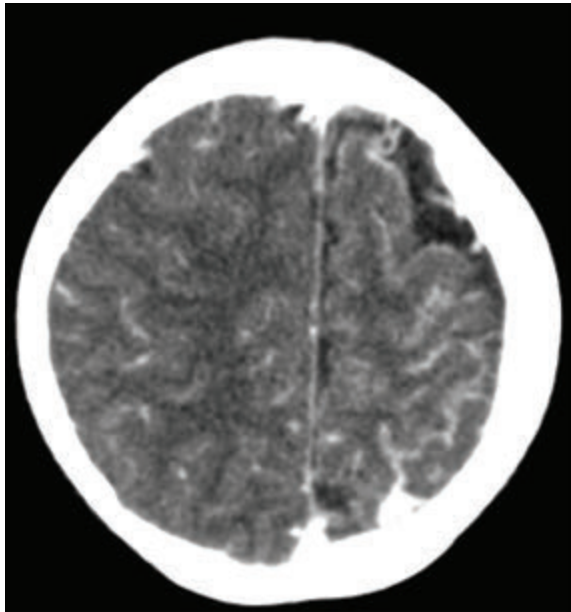
Fig. 3   Left cerebral hemispheric atrophy and subcortical gyral calcification along left cerebral convexity with angiomatous vascular malformation were revealed on computerized tomography.

Fig. 4   Computerized tomography demonstrated tram-track gyriform subcortical calcification involved right cerebral hemisphere. Right cerebral hemispheric atrophy with mild dilatation of ipsilateral the ventricular system was also detected.

patients who suffered from seizures could be treated with antiepileptic medication. The surgical treatment was performed in one patient with Phacomatosis pigmentovascularis type IIb. This patient was treated with functional hemispherectomy, temporal lobectomy, disconnection of white fiber at insular lobe, and falx cerebri. Although neurological impairment was found in 12 patients, neurological imaging was performed in the majority of cases (14 patients). Intracranial abnormalities were demonstrated in 11 patients (78.57%) including cerebral atrophy (81.82%), cerebral calcification (54.55%), leptomeningial angioma (27.27%), and enlarged choroidal plexus (27.27%). All of detected leptomeningial angioma involved the occipital lobes and posterior parietal lobe (Fig. 3, 4). The intracranial abnormality was also usually ipsilateral to the PWS. A patient with unilateral PWS, however, had bilateral intracranial lesion. Other systemic problems were detected in patients with Klippel-Trenaunay-Weber syndrome and Phacomatosis pigmentovascularis type IIb. Hypertension, adenoid hypertrophy, and severe obesity were found in a patient with Klippel-Trenaunay-Weber syndrome. Subglottic stenosis was demonstrated in a patient with Phacomatosis pigmentovascularis type IIb. Demographic data, clinical manifestation, treatments given, and neurological imaging were shown in Table 1.

Discussion

A child with Sturge-Weber syndrome (SWS) typically presents at birth with a facial cutaneous vascular malformation, port-wine stain (PWS). However, among patients who present with facial PWS, only 8 to 20% develop neurological symptoms[2-4]. The presence of PWS, therefore, is not sufficient to diagnose SWS. The authors, thus, excluded one patient with facial PWS who did not have ophthalmic or neurological evidence from the present study. On the contrary, some patients with SWS (5-15%) lack facial cutaneous vascular malformation[5]. SWS are significantly associated with PWS of the upper eyelid, bilateral topography of the PWS and unilateral PWS involving V1-3 area[6-8]. The risk of associated neurological and/or ocular disorder in a patient with partial or full V1 (forehead and/or eyelid) involvement was 18 to 65% in previous studies, glaucoma and epilepsy being the most common manifestations[1,7,8]. All of our patients had PWS in V1 area, accompany with neurological problem and/or ocular manifestation. However, seizure and glaucoma were the most common neurological and ocular disorder detected in

Table 1. Data of patients with Sturge-Weber syndrome

No.	Age	Sex	PWS	V area	Neuro. signs	Seizure onset	Rx seizure	Neurological imaging	Ocular signs	Glaucoma onset	Glaucoma Rx	F/U (year)
1	5 months	F	L	1, 2	Seizure Hemiparesis	5 months	Phenobarbital Phenyltoin	Cerebral atrophy Calcification	Dilated episcleral vss, glaucoma	12 years	Timolol Dorzolamide Bimatoprost	19
2	24 years	F	L	1, 2	Seizure Hemiparesis	2 years	Phenobarbital Phenyltoin	-	-	-	-	20
3	35 years	F	R	1, 2	Headache Seizure	44 years	Phenyltoin Depakine	Calcification	Dilated episcleral vss, glaucoma	10 years	Trabeculectomy Cyclodestruction	12
4	2 years	M	R	1, 2 (radiation)	-	-	-	Normal	Dilated episcleral vss, glaucoma bl in Schlemm's canal	8 years	Timolol Latanoprost	11
5	At birth	M	L	1, 2	-	-	-	Normal	Congenital glaucoma Megalocornea, corneal haziness	At birth	Trabeculectomy	12
6	8 months (Kippel Trenaunay Weber syndrome)	M	R	1, 2, 3	Seizure Delayed development	2 months	Phenobarbital	Cerebral atrophy Calcification Enlarged plexus	Congenital glaucoma, buphthalmos Dilated episcleral vss	2 months	Brinzolamide Bimatoprost	11
7	1 month	F	R	1, 2	-	-	-	Normal	Ptosis, anisometropia Amblyopia, strabismus	-	-	7
8	6 months	F	L	1	Seizure Hemiparesis	4 months	Toparimate Levetiracetam	Cerebral atrophy Enlarged plexus	No ocular exam.	-	-	7
9	1 year	R L	2 1, 2	M	Seizure, hemiparesis Delayed development Hearing loss	5 months	Phenyltoin	Bilateral cerebral atrophy	No ocular exam.	-	-	2
10	8 months	M	L	1, 2, 3 (Laser Rx)	Seizure	8 months	Depakine	Bilateral cerebral atrophy	Glaucoma, choroidal hemangioma	2 years	Timolol	8.5
11	3 months (Phacomatosis pigmentovascularis: IIb)	M	R L	1, 2, 3 3	Delayed development Seizure Hemiparesis	2 months	Surgery Phenobarbital Levetiracetam	Cerebral atrophy Enlarged plexus	Congenital glaucoma, buphthalmos BE Dilated episcleral & tortuous retinal vss BE bl in Schlemm's canal, corneal haziness RE	1 month	Brinzolamide Latanoprost Cyclodestruction	8
12	5 years	F	R	1, 2	Seizure Hemiparesis	3 months	Phenobarbital Phenyltoin	Cerebral atrophy Calcification	-	-	-	2.5
13	8 years	M	R L	1, 2, 3 1, 2, 3	Seizure Headache	6 months	Phenyltoin	Cerebral atrophy Calcification, angioma	Megalocornea, dilated episcleral vss BE Choroidal hemangioma, glaucoma BE	8 years	Timolol	8
14	3 months	M	R L	1, 2 2	Seizure	3 months	Depakine Phenobarbital Carbamazepine	Angioma	No ocular exam.	-	-	2.5
15	11 months	F	R	1, 2, 3	Seizure	11 months	Depakine	Cerebral atrophy Calcification, angioma	-	-	-	4.5
16	31 years	M	R	1, 2, 3	-	-	-	-	Dilated episcleral vss, glaucoma Amblyopia, strabismus Choroidal hemangioma	3 years	-	0.5

PWS = port-wine stain; V area = trigeminal nerve branch area; M = male; F = female; R = right; L = left; vss = vessel; bl = blood; RE = right eye; BE = both eyes

this retrospective study. In a literature review, the involvement of V1 alone is up to 20% in patient with SWS and bilateral PWS lesions can be seen in 10 to 30% of the patients[1]. Although bilateral PWS was detected in 25% of the patients in the present study, most patients (93.75%) had PWS extending beyond the V1 facial area. PWS can be treated with pulsed dye laser in the same way that is treated in children without SWS. Unfortunately, only two patients in the present study were treated.

Ocular involvements in SWS arise primarily from vascular abnormalities of the conjunctiva, episclera, retina, and choroid. Ocular manifestations of SWS were episcleral/conjunctival hemangioma, iris heterochromia, glaucoma, choroidal hemangioma, optic disc coloboma, and cataract[1]. The ocular complication is usually ipsilateral to the PWS. However, the authors found the ocular problem in both eyes of a patient who had PWS in V1-3 on the right side and only V3 facial area on the other side. Glaucoma is the most common manifestation, an incidence of 60 to 74.5% has been previously reported[8-11], and this occurred in 56.25% of our patients. The onset of glaucoma is bimodal. From a literature review, about 60% of patients develop glaucoma in infancy when the eye is susceptible to increased intraocular pressure and 40% in childhood or later[5]. The authors found 55.56% of our patients develop glaucoma before three years of age. Only 33.33% of studied patients develop glaucoma in infancy. The main mechanism underlying glaucoma is the increasing resistance to aqueous fluid outflow. Early onset of glaucoma is typically due to vascular abnormalities in the anterior chamber angle and outflow tract causes infants to develop corneal abnormalities including megalocornea, buphthalmos, and corneal haziness. Late-onset glaucoma may not occur until adolescence. It is usually the result of increase in episcleral venous pressure caused by episcleral arteriovenous shunts, which prompt little to no corneal enlargement. The study conducted by Shiau et al, using the episcleral venomanometer, supports this hypothesis[12]. Neely D and co-workers investigated the ocular hemodynamics of SWS associated glaucoma using color Doppler imaging techniques. These investigators did not find arterial retrobulbar blood flow differences between the glaucomatous and the fellow normal eye since the primary vascular anomaly was in the venous plexus[13].

There was only a single case report in the literature with acute glaucoma secondary to posterior scleritis in SWS[14]. Although the corneal abnormalities usually associated with infantile glaucoma, one of our SWS patients with bilateral extensive PWS and diffuse choroidal hemangioma had bilateral megalocornea with late-onset glaucoma diagnosis. The only reliable test that can be done during the follow-up period was measurement of intraocular pressure, because visual field testing was very difficult in young children. In some cases, control of glaucoma was difficult. Medical treatment, however, should be considered as the initial treatment of choice[15-17]. Prostaglandin analogues should be used with caution because it can induce uveal effusion[18,19]. Different surgical procedures have been performed in uncontrolled cases including cyclodestructive procedure, goniotomy, and trabeculectomy. Molteno or Ahmed implants may be necessary if these fail or even as the first intervention[20]. Although the maximum dose of topical antiglacomatous drug was used for our patients, the surgical procedures were needed in four eyes. Unfortunately, four eyes with infantile glaucoma in the present study went blind.

Diffuse choroidal hemangioma was another characteristic ophthalmic finding of SWS because choroidal vessels also exhibit angiomatous changes that may be found in up to 71% of SWS cases (24.4-71%)[1,7,9,21,22], but occurred in only 18.75% of our patients. It is usually ipsilateral to PWS as noticed in the present study. Amirikia et al, however, reported a 6-year-old male who had unilateral PWS with bilateral choroidal hemangioma[23]. Diffuse choroidal hemangioma is usually seen as a red, flat to moderately elevated mass-producing a classic "tomato ketchup" appearance. These lesions can initiate progressive secondary changes in the overlying retina such as retinal pigment degeneration, fibrous metaplasia, and cystic retinal degeneration leading to visual loss and visual field defect. B-scan ultrasonography could demonstrate the diffuse thickening of the choroid (Fig. 5), and other posterior segment complications in case with media opacity especially infantile glaucoma with corneal complication. Visual loss may result from refractive error, foveal distortion, as well as exudative retinal detachment as seen in one of our patients. Optional treatments of serous retinal detachment secondary to the diffuse choroidal hemangioma include external beam radiation, proton beam therapy, brachytherapy, photodynamic therapy, and possibly anti-VEGF. Although the photodynamic therapy (PDT) has theoretical properties for treating circumscribed choroidal hemangioma, to date, only four cases of
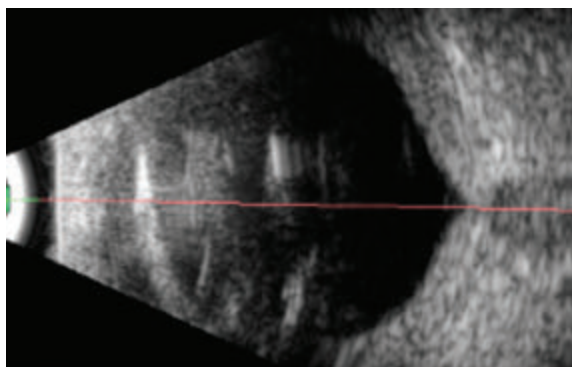
Fig. 5 B-scan ocular ultrasonography shows diffuse thickening of choroid which compatible with diffuse choroidal hemangioma.

diffuse choroidal hemangioma treated with PDT have been published. Most authors agree that avoiding overlapping PDT spots is prudent because of the theoretical risk. Likewise, treatment of fovea is avoided to reduce scarring and pigmentary changes[24].

Patients with SWS suffer from variety of neurologic abnormalities, including epilepsy, hemiparesis, mental retardation, attention-deficit hyperactivity disorder, migraine, headache, and stroke-like episodes. Twenty three to 89% of children with SWS have convulsion[1,11,25]. Convulsion was found in 75% of our patients. The age of presentation of convulsion and their evolution is variable[5]. In a retrospective case series, the onset of seizure ranged from birth to 23 years, and 12% of patients did not develop seizure until the third decade of life[10]. Focal motor seizures are the most common manifestation because the lesion responsible for the epilepsy in SWS patients with neurological impairment is focal, usually present by the age of three years. Children whose seizures begin before the age of two years or intractable seizure with anti-epileptic drug may eventually lead to motor deficits and mental retardation[25,26], and this occurred in 30% of our patients. Global delayed development was detected in two patients who were diagnosed with Klippel-Trenaunay-Weber syndrome and Phacomatosis pigmentovascularis type IIb.

Fortunately, Most of our patient who presented with seizure could be controlled with anti-epileptic drug. The role of prophylactic anti-epileptic drug and low dose aspirin medications remains controversial[1]. Phenyltoin-induced gingival hypertrophy may aggravate periodontal and dental problems, as noticed in one patient of this retrospective study. Topiramate should be used with caution as it

can cause acute myopia and secondary angle closure glaucoma in a small subset of people who take topiramate regularly[27]. Intractable seizure was demonstrated, in the present study, in one patient with Phacomatosis pigmentovascularis type IIb and surgical intervention was given.

Headaches affect 30 to 60% of patient with SWS[25,28]. The present study, however, we found only 12.5% of patients had headaches. This may result from the small number of cases collected in the present study or the rather young age of patients studied. Leptomeningeal angioma may predispose them to neuronal hyperexcitability, causing changes in cortical perfusion and oxygenation consistent with theories on pathogenesis of migraine[29]. Transient focal neurological deficits or stroke-like episodes are a unique feature in SWS, with the most common manifestation being transient episodes of hemiparesis or visual field defects not directly associated with epilepsy. These episodes are frequently more prolonged than a postictal paresis and may last several days, weeks, months, or become permanent. Importantly (in contrast to Todd's palsy), stroke-like episode often precedes the onset of seizure. Recurrent thrombosis is a hypothetical mechanism of stroke-like episode[5,25]. So, hemiparesis was observed in 25 to 60% of patient with SWS[25,30,31], as revealed in 37.5% of our patients. Whether the prevention of stroke-like episodes improves outcome is unknown. A few studies showed the benefit of low-dose aspirin, reduction in both seizure frequency and stroke-like episodes, although the bleeding complications were reported in some SWS patients[32,33].

The intracranial leptomeningeal angiomatosis is a key diagnostic feature in SWS, it may not be apparent early in infancy. The longitudinal studies have not yet been undertaken to define the optimal age of neuroimaging screening. The characteristic radiologic features are vascular abnormalities that commonly located in the parieto-occipital area. Both cerebral atrophy and cortical calcifications are considered to be an indirect consequence of chronic ischemia of the cortex due to vascular stasis in the area of leptomeningeal angioma[1]. Plain skull X-rays illustrate the classic "tram-line" or "tram-track" calcification but are helpful only in diagnosis of SWS late in life. Angiography is no longer used to visualize the vascular abnormalities because they can be observed more clearly and non-invasively with magnetic resonance imaging (MRI)[25]. Computed tomography provides adequate evaluation of brain calcification. However,

calcifications may be absent or minimal in infant. Therefore, MRI with contrast is the preferred and most sensitive imaging technique for evaluation of the leptomeningeal angiomatosis[1,25]. The incidence of neurological abnormalities in SWS is higher among patients with bilateral PWS and bilateral intracranial involvement is uncommon even in patients with bilateral PWS. The authors conversely found bilateral intracranial lesions in a patient with unilateral extensive PWS, and unilateral intracranial lesion in a patient with bilateral extensive PWS. The imaging may also show thickening of the ocular globe as a manifestation of diffuse choroidal hemangioma especially in unilateral cases which the opposite side can be used to compare. At present, metabolic imaging studies may also assist in characterizing the extent of brain disease in SWS[5,33].

The cause of SWS is still unknown; however, non-Mendelian genetic hypotheses have been suggested. Comi AM and co-workers reviewed the dysmorphic features, structural abnormalities, and masses seen in patient with SWS. They revealed a number of genes in the 17p1-p13 region with links to the unexpected abnormalities[34]. Many unexpected abnormalities were previously reported such as cerebral astrocytoma, retinitis pigmentosa, deafness, subglottic stenosis, and inguinal hernia. Many syndromes associated with SWS had ever been published; such as Usher syndrome, Klippel-Trenaunay syndrome and Phacomatosis pigmentovascularis type IIb[35-39]. Further research is needed to evaluate the causing genes. Furthermore, for patients with SWS and associated unusual findings, it may appropriate to perform genetic testing.

Conclusion

Port-wine stains, glaucoma, and epilepsy were the most frequent clinical features of patients with SWS in the present study. When a child is born with PWS covering the facial area of V1 distribution, MRI should be performed to screen for leptomeningeal angiomatosis and complete ocular examination should be carried out to screen for ocular complication especially glaucoma. Multidisciplinary team management as well as life-long follow-up is required.

What is already known on this topic?

Sturge-Weber syndrome (SWS) is a rare sporadic neurocutaneous syndrome. Although SWS is among the common phakomatosis, the pathogenesis remains unclear and no true animal model exists. The clinical manifestations of SWS are varied widely, with absent or varying neurological and/or ocular features in some cases.

What this study adds?

This study provided the additional formation of SWS including clinical manifestation, neuroimaging, and management strategy. We also reported two rare associated syndrome, Klippel Trenaunay syndrome, and Phacomatosis pigmentovascularis.

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การศึกษาแบบย้อนหลัง 12 ปีของกลุ่มอาการ Sturge-Weber และการทบทวนวรรณกรรม

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วัตถุประสงค์: เพื่อศึกษาทบทวนอาการแสดงทางคลินิกและผลการตรวจทางรังสีวิทยาของระบบประสาทของผู้ป่วยกลุ่มอาการ Sturge-Weber ที่มารับการรักษา ณ โรงพยาบาลศรีนครินทร์ ตลอดระยะเวลา 12 ปี

วัสดุและวิธีการ: การศึกษาแบบย้อนหลังในผู้ป่วยกลุ่มอาการ Sturge-Weber จำนวน 16 ราย (เพศชาย 9 ราย และเพศหญิง 7 ราย) โดยทำการทบทวนข้อมูลเวชระเบียน ภาพถ่ายต่างๆ และผลการตรวจทางรังสีวิทยาของระบบประสาท

ผลการศึกษา: ผู้ป่วยทุกรายมี port-wine stain (PWS) ในบริเวณเปลือกตา โดยผู้ป่วยจำนวน 4 ราย (25%) มีรอยโรคที่ผิวหนังบนใบหน้าทั้งสองข้าง โรคต้อหินถูกพบเป็นโรคทางจักษุวิทยาหลัก ผู้ป่วยจำนวน 9 ราย (56.25%) ได้รับการวินิจฉัยโรคต้อหิน 11 ตา โดยมีจำนวน 4 ตา ที่จำเป็นต้องรับการรักษาด้วย cyclodestructive procedure และหรือการผ่าตัด ในท้ายที่สุดพบมีตาบอดจำนวน 4 ตา ความผิดปกติทางจักษุวิทยาอื่นๆ ได้แก่ ภาวะสายตาผิดปกติ หลอดเลือด episcleral ขยายตัว กระจกตาผิดปกติ หลอดเลือดจอตาคดงอ choroidal hemangioma ภาวะตาขี้เกียจ และตาเหล่ ผู้ป่วยจำนวน 12 ราย (75%) มีความบกพร่องทางระบบประสาท ได้แก่ ชัก อัมพฤกษ์ ปวดศีรษะและมีพัฒนาการช้า อย่างไรก็ตาม อาการแสดงทางระบบประสาทที่พบบ่อยที่สุดคือการชัก (75%) ซึ่งสามารถควบคุมได้ด้วยยากันชัก ผู้ป่วยส่วนใหญ่ (14 ราย) ได้รับการตรวจทางรังสีวิทยาของระบบประสาท โดยพบความผิดปกติในผู้ป่วยจำนวน 11 ราย (78.57%) ได้แก่ สมองฝ่อ (81.82%) หินปูนในสมอง (54.55%) leptomeningeal angioma (27.27%) และการขยายขนาดของ choroidal plexus (27.27%) สำหรับความผิดปกติของดวงตาและสมองนั้นมักพบในข้างเดียวกับรอยโรคที่ผิวหนัง (PWS) อย่างไรก็ตาม ผู้ป่วยรายหนึ่งพบรอยโรคในสมองทั้งสองข้างทั้งที่มี PWS เพียงข้างเดียว

สรุป: การศึกษานี้พบว่า port-wine stain โรคต้อหิน และโรคชัก เป็นลักษณะเด่นทางคลินิกของกลุ่มอาการ Sturge-Weber ซึ่งผู้ป่วยควรได้รับการตรวจประเมินทางจักษุวิทยาและระบบประสาทอย่างละเอียดตั้งแต่ได้รับการวินิจฉัยโรค ผู้ป่วยมีความจำเป็นต้องได้รับการดูแลรักษาโดยกลุ่มผู้เชี่ยวชาญสหสาขาวิชาตลอดจนการติดตามอาการไปตลอดชีวิต