

# Severe Sturge-Weber syndrome in a 9-year-old boy: a great challenge

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Dear Editor,

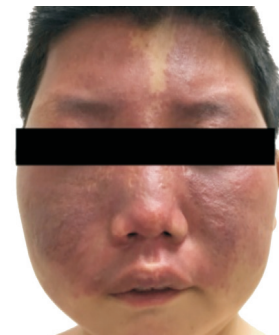
We present a severe Sturge-Weber syndrome (SWS) in a boy with Mongolian spot, congenital glaucoma, seizures, and intelligent disability. During the hospital, the boy was admitted Pediatrics Department and Ophthalmology Department to control the seizure of episode and glaucoma respectively. This case demonstrates the diagnosis and treatment process of complex systematic disease and the implications of the treatment process. The diagnosis and treatment conformed to the Declaration of Helsinki, and the family members gave written informed consent.

SWS is a congenital vascular disorder characterized by facial port-wine stains distributed along the trigeminal nerve, which is complicated with various ophthalmic, neurological, and cognitive symptoms of variable severity<sup>[1-2]</sup>.

The boy with a large number of 'red birthmarks' came to the clinic because of a decline in vision (Figure 1). He is diagnosed with epilepsy and mental retardation since he was 6 years old. Systematic physical examination showed that facial port-wine stains were distributed along the 3 branches of the trigeminal nerve and were also presented over the right arm, chest and back. Besides, the boy was found greyish blue patches (Mongolian spots) spreading over the chest, back, abdomen, waist, buttocks, and thigh (Figure 2). Physical examination

of his eyes was not cooperative. Intraocular pressure (IOP) was 37 mm Hg in the right eye and 34 mm Hg in the left eye. Cornea showed post elastic layer fracture. Its diameter was 12 mm in both eyes. On examination of the fundus, cup-to-disc area ratio (C/D) was 1.0 in the both eyes (Figure 3).

Optic coherent tomography (OCT) scans revealed that the choroid baseline in the macular area was flat, the choroid capillary layer was thicker, and the retina was thinner (Figure 4). Fundus fluorescein angiography (FFA) revealed that diffuse choroidal hemangioma. Magnetic resonance imaging (MRI) showed meningeal abnormal enhancement and an arc-shaped abnormal signal in the posterior pole of the bilateral ring plates. The arc-shaped showed low signal in both T1- and



**Figure 1** 'Red birthmarks' on the face Facial port-wine stains distributed along the trigeminal nerve.



**Figure 2** Skin lesion A: Front picture; B: Back picture. Arrow sign showing greyish blue patches and triangle sign showing port-wine stains.

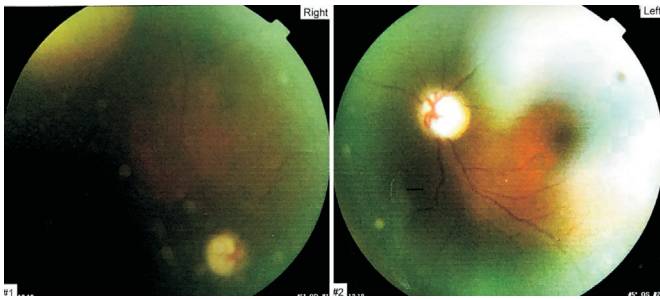


Figure 3 Fundus picture C/D=1.0 in both eyes.

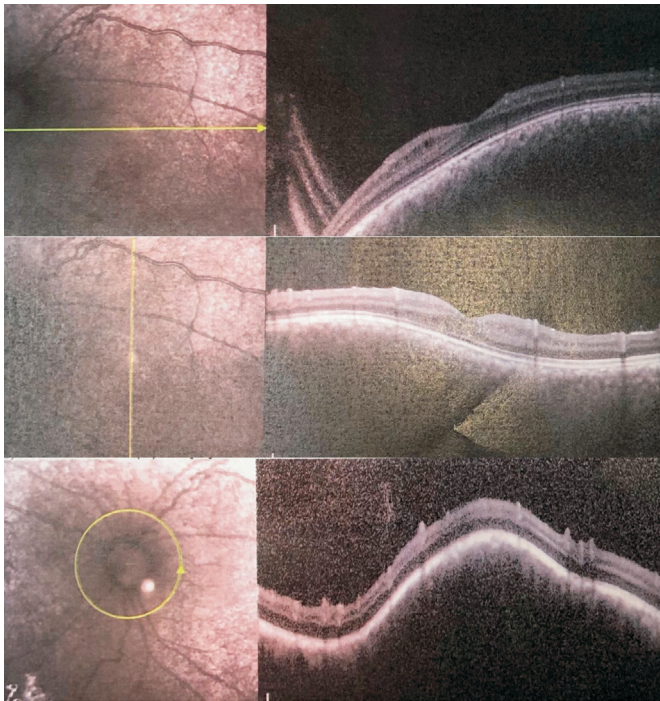


Figure 4 OCT image.

T2-weighted images, and obvious and uniform enhancement in enhanced scanning. The magnetic resonance venography (MRV) demonstrated multiple varicose and tortuous veins at the bilateral iliac crest and the left occipital plexus. It was filling defect in the superior sagittal sinus, the inferior sagittal sinus, the left transverse sinus, and the upper end of the internal jugular vein (Figure 5). No abnormalities were found in the results of full blood tests, serum chemistries, electrocardiograms, cardiac and abdominal Doppler ultrasound, or chest X-rays. With the limit of cognitive function, the patient cannot cooperate with some examinations, so that some necessary examinations, such as MRI, MRV, were performed under sedation. To ensure the accuracy of the IOP, the IOP were all measured by Goldman tonometer when the patient was awake.

In terms of treatment, we must first control epilepsy. After controlling epilepsy, Ahmed valve implantation combined with allogeneic scleral flap implantation in his right eye was performed under general anesthesia (Figure 6). However, on the third day after the operation, the IOP of the right eye

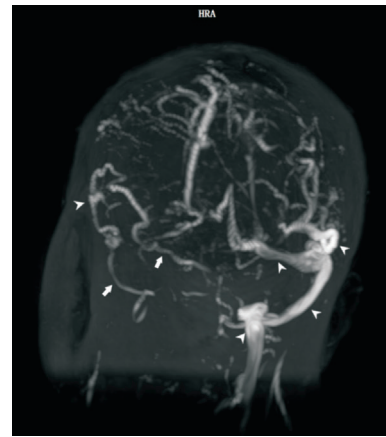


Figure 5 Brain MRV image Arrow sign showing narrow veins and triangle sign showing dilated veins.

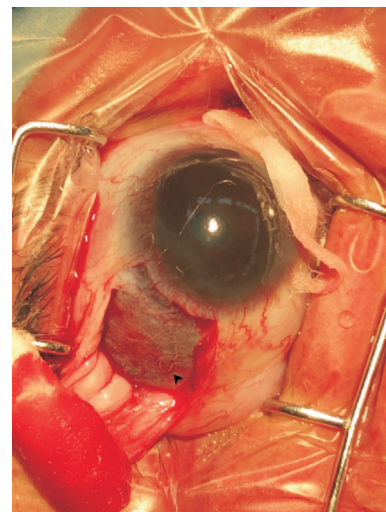


Figure 6 Picture in operation Triangle sign showing diffuse choroidal hemangioma and the thinned sclera.

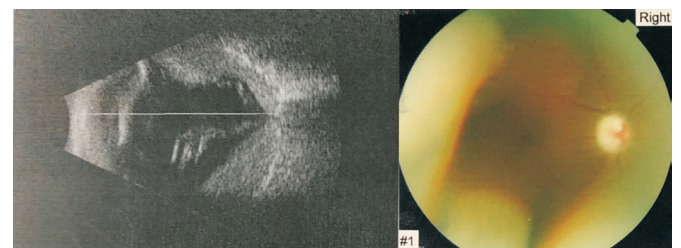


Figure 7 B-ultrasound and fundus picture Both pictures showing retinal detachment in the right eye.

was 7 mm Hg and anterior chamber disappeared with the tip of the tube attached to the endothelium. At the same time, B-ultrasound also revealed retinal detachment in the right eye (Figure 7). Therefore, three anterior chamber reconstructions were performed. Due to the high risk and cost of the surgery, the family decided to abandon the surgery in the left eye. To control the IOP in the left eye, we decided to switch to drug therapy with multiple compound preparations. During the follow-up from discharge to August 2019, the IOP fluctuated 16-23 mm Hg in the right eye and 20-40 mm Hg in the left eye with 3 drugs.

To our knowledge, SWS is a malformation of the blood vessels involving the skin, eyes, and brain. Vascular malformations can cause glaucoma. The two main theories of pathophysiology are malformation of the anterior chamber angle and increased episcleral venous pressure<sup>[3]</sup>. For many anatomical anomalies of this glaucoma are similar to those found in congenital glaucoma, malformation of the anterior chamber angle may be the cause of early-onset glaucoma, while elevated episcleral vein pressure may play a greater role in late-onset glaucoma. Infantile glaucoma is manifested by increased vascularity of the conjunctiva, buphthalmos, or increased tearing<sup>[4]</sup>. Children with a port-wine birthmark (PWB) on the upper and lower eyelids are at high risk for glaucoma, who is required to detect glaucoma and initiate treatment every few months in infancy and early childhood.

Glaucoma is treated with eye drops to reduce IOP at early stage<sup>[5]</sup>. When medical therapy is unsuccessful or glaucoma has reached the advanced stage, filtration surgery is required<sup>[6]</sup>. However, regardless of the surgical method, the possibility of complications after surgery is greater, especially in trabeculectomy<sup>[5,7]</sup>. In our case, Ahmed valve implantation combined with allogeneic scleral flap implantation in the right eye was performed. Although patients had complications of retinal detachment in a short time, it all improved after treatment. The possible causes of complications are: first, the patient is in the advanced stage of the disease and has multiple clinical manifestations; second, the inevitable repeated rubbing of the eyes led to hypotonia and shallow of the anterior chamber.

Imaging examination plays an important role in the diagnosis of brain involvement. Although contrast-enhanced MRI has decreased sensitivity in young infants, susceptibility-weighted imaging and postcontrast flair sequences may increase sensitivity. Therefore, image examination must be repeated after a year of age<sup>[8-9]</sup>.

Method of visualizing the enhancing leptomeningeal vessels, such like MRV may be helpful in brain involvement. In our case, MRV showed the shape of veins and the internal jugular veins were also visualized. Contrast-enhanced MRI provides enough evidence for diagnosing brain involvement, but MRV provides more intuitive results.

Epilepsy is a serious problem that requires great attention in children with SWS. Approximately 15% of patients develop the disease within 1 year of age and 90% of patients develop the disease within 2 years of age<sup>[10]</sup>. Also, seizure onset before age 1y has a crucial effect on the severity of cognitive and motor dysfunction in children with SWS<sup>[11]</sup>. The most commonly used anticonvulsants in infants include oxcarbazepine or carbamazepine, and levetiracetam<sup>[12]</sup>. Most patients can achieve reasonable seizure control with 1 or 2

anticonvulsants and low-dose aspirin<sup>[13]</sup>. As a result, anti-epileptic treatment should be actively performed.

Currently, the role of the GNAQ test in diagnosis remains to be determined. It is very regrettable in our case that the corresponding tissue was not obtained for GNAQ testing.

#### ACKNOWLEDGEMENTS

**Conflicts of Interest:** Pan Z, None; Chen HH, None; Jiang B, None; Shi JM, None.

#### REFERENCES

- 1 Shirley MD, Tang H, Gallione CJ, *et al.* Sturge-Weber syndrome and port-wine stains caused by somatic mutation in GNAQ. *N Engl J Med* 2013;368(21):1971-1979.
- 2 Cho S, Maharathi B, Ball KL, Loeb JA, Pevsner J. Sturge-weber syndrome patient registry: delayed diagnosis and poor seizure control. *J Pediatr* 2019;215:158-163.e6.
- 3 Thavikulwat AT, Edward DP, AlDarrab A, Vajaranant TS. Pathophysiology and management of glaucoma associated with phakomatoses. *J Neurosci Res* 2019;97(1):57-69.
- 4 Mantelli F, Bruscolini A, la Cava M, Abdolrahimzadeh S, Lambiase A. Ocular manifestations of Sturge-Weber syndrome: pathogenesis, diagnosis, and management. *Clin Ophthalmol* 2016;10:871-878.
- 5 Iwach AG, Hoskins HD Jr, Hetherington J Jr, Shaffer RN. Analysis of surgical and medical management of glaucoma in Sturge-Weber syndrome. *Ophthalmology* 1990;97(7):904-909.
- 6 Patrianakos TD, Nagao K, Walton DS. Surgical management of glaucoma with the sturge weber syndrome. *Int Ophthalmol Clin* 2008;48(2):63-78.
- 7 Hamush NG, Coleman AL, Wilson MR. Ahmed glaucoma valve implant for management of glaucoma in Sturge-Weber syndrome. *Am J Ophthalmol* 1999;128(6):758-760.
- 8 Porto L, Kieslich M, Yan B, Zanella FE, Lanfermann H. Accelerated myelination associated with venous congestion. *Eur Radiol* 2006;16(4):922-926.
- 9 Fischbein NJ, Barkovich AJ, Wu Y, Berg BO. Sturge-Weber syndrome with no leptomeningeal enhancement on MRI. *Neuroradiology* 1998;40(3):177-180.
- 10 Sujansky E, Conradi S. Sturge-Weber syndrome: age of onset of seizures and glaucoma and the prognosis for affected children. *J Child Neurol* 1995;10(1):49-58.
- 11 Luat AF, Behen ME, Chugani HT, Juhász C. Cognitive and motor outcomes in children with unilateral Sturge-Weber syndrome: Effect of age at seizure onset and side of brain involvement. *Epilepsy Behav* 2018;80:202-207.
- 12 Kaplan EH, Kossoff EH, Bachur CD, Gholston M, Hahn J, Widlus M, Comi AM. Anticonvulsant Efficacy in Sturge-Weber Syndrome. *Pediatr Neurol* 2016;58:31-36.
- 13 Lance EI, Sreenivasan AK, Zabel TA, Kossoff EH, Comi AM. Aspirin use in Sturge-Weber syndrome: side effects and clinical outcomes. *J Child Neurol* 2013;28(2):213-218.