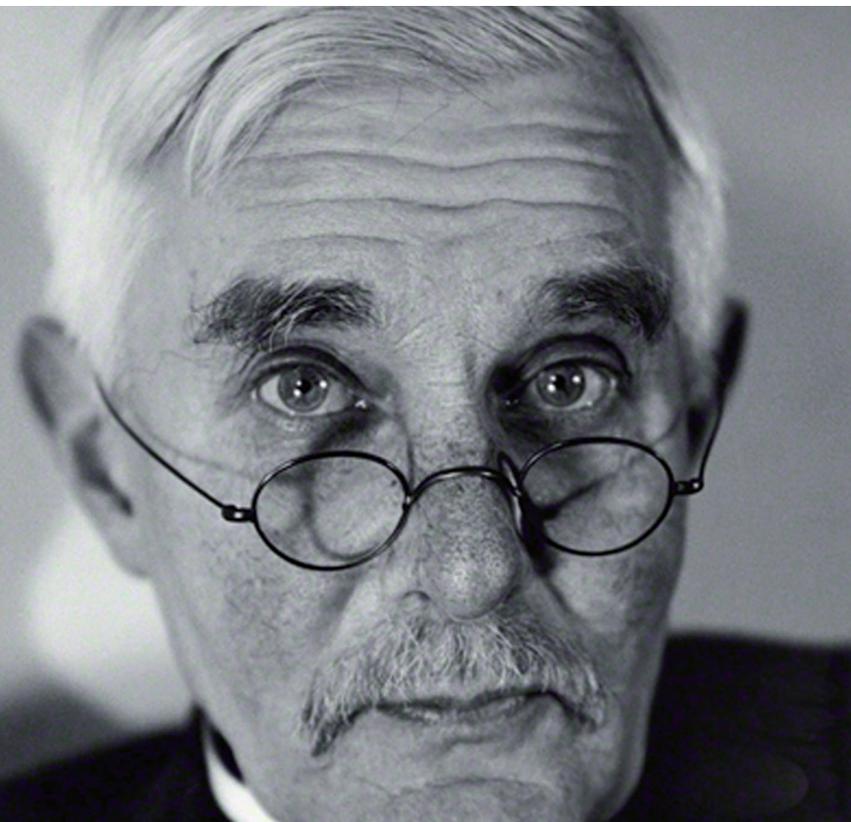


SWISS SOCIETY OF NEONATOLOGY

Sturge Weber syndrome,
phakomatosis pigmentovascularis and Down syndrome
in a newborn infant

January 2014



Sturge Weber syndrome (SWS) is a neurocutaneous disorder affecting skin, central nervous system and eyes. Phakomatosis pigmentovascularis (PPV) is another neurocutaneous disorder with an association of cutaneous and extracutaneous (visceral, muscular, neurologic or ocular) abnormalities.

We present a newborn African boy with SWS with port wine stains and leptomeningeal involvement. Additionally, he had multiple Mongolian spots and was also diagnosed with Down syndrome.

This boy was born to a 30-year old mother by spontaneous vaginal delivery at 41 3/7 weeks of gestation following premature and prolonged rupture of membranes (10 days). The prenatal ultrasound examinations had been unremarkable. Both parents were from Nigeria and there was no consanguinity.

CASE REPORT

In the delivery room, the infant was intubated because of poor respiratory effort. Apgar scores were 4, 6 and 7 at 1, 5 and 10 minutes, respectively. The arterial umbilical cord pH value was 7.21. He was transferred to our neonatal intensive care unit. Birth weight was 3640 g (P 25-50), body length 50 cm (P 5-10) and head circumference 35 cm (P 10-25). On physical examination, there were facial port wine stains on the left side in the distribution of the first and second branches of the trigeminal nerve (Fig. 1). Furthermore, he showed

the port wine stains and numerous Mongolian spots, some of them of huge size, all over his body (Fig. 2). Finally, there were stigmata of Down syndrome: low-set small ears, a simian crease, short fingers and a sandal gap. On the second day after birth, the boy was successfully extubated. Respiratory support with high flow and supplemental oxygen (maximum FiO_2 0.6) was required for nine days.

Chest X-ray was normal. Echocardiography in the first days of life revealed persisting pulmonary arterial hypertension with right-to-left shunting through the patent foramen ovale and the patent ductus arteriosus, but no structural anomalies. Cranial ultrasound showed no pathological findings. The cranial MRI revealed left-sided meningeal angiomas and an accelerated myelination (Fig. 3). Ophthalmologic examination was normal without vascular malformation or signs of glaucoma. On abdominal ultrasound, there was increased echogenicity of the kidneys, but no signs of malformation. Chromosomal analysis confirmed Down syndrome. The port wine stains and meningeal angiomas were felt to be compatible with SWS. The combination of port wine stains with Mongolian spots is characteristic of phakomatosis pigmentovascularis.

Apart from muscular hypotonia, the boy showed no noticeable neurological problems (e.g., seizures) during



Fig. 1

Typical facial involvement of Sturge Weber syndrome in the distribution of the trigeminal nerve.



Fig. 2

Large port wine stains and Mongolian spots all over the body.



his hospitalisation. He was discharged at the age of 14 days and regular hematological and neurological follow-ups were scheduled. Multiple episodes of viral upper respiratory tract infections led to rehospitalizations for supplemental oxygen and gavage feedings.

Low dose aspirin (5 mg/kg/d) was recommended by the neurologists and started at the age of 3 months. It is assumed that prophylactic anticoagulant therapy in children with SWS prevents the progression of impaired cerebral blood flow and hypoxic-ischemic neuronal injury. Up to the age of 5 months, the patient was free of seizures and his neurological examination was normal except for some mild muscular hypotonia.

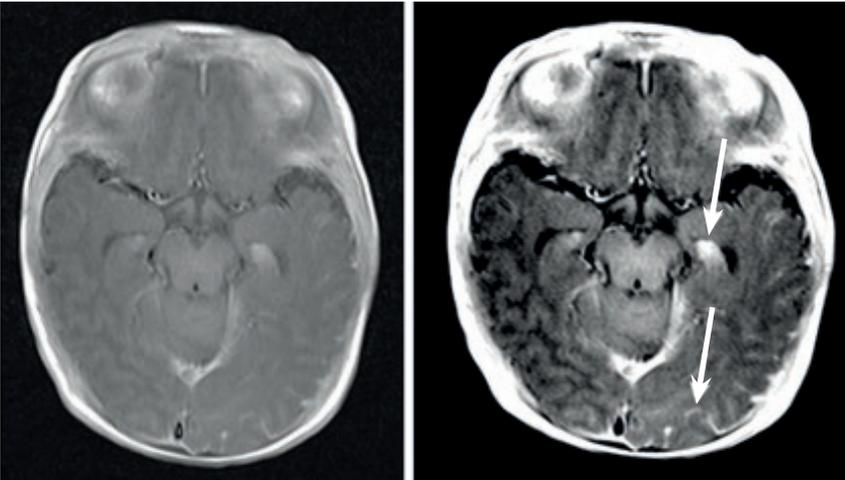


Fig. 3

Cranial MRI (axial T1-weighted image with gadolinium): occipital leptomenigeal angiomas (lower arrow).

DISCUSSION

Sturge Weber syndrome (SWS) is a rare congenital vascular disease that occurs sporadically (1:40'000-50'000 live births). The etiology is unknown, but it is not a heritable disorder (1). It typically presents with facial capillary malformations - known as port wine stains - in the distribution of the trigeminal nerve, often bilateral. The clinical diagnosis is based on the presence of a classical triad: facial capillary malformations, leptomeningeal angiomatosis and glaucoma (2).

If the port wine stains involve the forehead or the eyelid, involvement of the brain occurs in 10% to 20%, usually on the same side (3). Eighty percent of patients with SWS develop seizures: three quarters occur in the first year of life, and the remainder manifests usually by the age of five years. Initially, they are typically focal, but often become generalized tonic-clonic and can be associated with stroke-like symptoms such as acute hemiparesis or visual field defects. Seizure control can be challenging (2, 4).

Thirty to seventy percent of the patients with SWS develop glaucoma with the highest risk in the first ten years of life. Half of the patients present with congenital glaucoma (5, 6). In the first months of life, children with SWS show typically a normal development. Mental retardation occurs in half of the patients over time. In one third, there is severe developmental delay (7).

Phacomatosis pigmentovascularis (PPV) is another rare neurocutaneous disorder with vascular malformations and dermal melanocytosis. Multiple types of PPV have been identified. The most common type (85%) is the combination of port wine stains and blue spots like Mongolian spots or nevus of Ota (type II). In half of the patients, PPV type II is associated with systemic involvement, e.g. neurological, ocular, skeletal or renal abnormalities (8).

Mongolian spots represent a congenital dermal melanocytosis and are very common in black neonates (>60%). They are a completely benign result of delayed disappearance of dermal melanocytes and usually disappear during the first two years of life. The most common locations are the sacrogluteal region as well as the shoulders. Mongolian spots rarely occur on the head, face or extremities (9).

Our patient showed multiple port wine stains at typical and also uncommon locations and of unusual sizes. Additionally, the leptomeningeal angiomatosis supports our diagnosis of SWS. Considering the widespread capillary malformations and multiple Mongolian spots, a PPV type II can also be diagnosed. In our case, PPV type II and SWS are overlapping entities, since PPV type II can affect the central nervous system and the eyes as well. An association of SWS and PPV, as well as SWS and Down syndrome has previously been reported in the literature (10). However,

after a thorough search of the medical literature, this appears to be the only case of a combined manifestation of all three disorders.

1. Comi AM. Presentation, diagnosis, pathophysiology and treatment of the neurological features of Sturge-Weber-syndrome. *Neurologist* 2011;17:179-184
2. Bodensteiner JB, Roach ES. Sturge-Weber syndrome: Introduction and overview. In: *Sturge-Weber Syndrome*, 2nd edition, Bodensteiner JB, Roach ES (Eds), Sturge-Weber Foundation, Mt. Freedom, NJ 2010
3. Tallman B, Tan OT, Morelli JG, et al. Location of port-wine stains and the likelihood of ophthalmic and/or central nervous system complications. *Pediatrics* 1991;87:323-327
4. Sujansky E, Conradi S. Sturge-Weber syndrome: age of onset of seizures and glaucoma and the prognosis for affected children. *J Child Neurol* 1995;10:49-58
5. Sullivan TJ, Clarke MP, Morin JD. The ocular manifestations of the Sturge-Weber syndrome. *J Pediatr Ophthalmol Strabismus* 1992;29:349-56
6. Henedige AA, Quaba AA, Al-Nakib K. Sturge-Weber syndrome and dermatomal facial port-wine stains: incidence, association with glaucoma, and pulsed tunable dye laser treatment effectiveness. *Plast Reconstr Surg* 2008;121:1173-1180
7. Sujansky E, Conradi S. Outcome of Sturge-Weber syndrome in 52 adults. *Am J Med Genet* 1995;57:35-45
8. Fernandes-Guarino M, Boixeda P, de Las Heras E, et al. Phakomatosis pigmentovascularis: Clinical findings in 15 patients and review of the literature. *J Am Acad Dermatol* 2008;58:88-93
9. Cordova A. The Mongolian spot: a study of ethnic differences and a literature review. *Clin Pediatr (Phila)* 1981;20:714-719
10. Al Robaee A, Banka N, Alfadley A. Phakomatosis pigmentovascularis type IIb associated with Sturge-Weber syndrome. *Pediatr Dermatol* 2004;21:642-645

SUPPORTED BY



CONTACT Swiss Society of Neonatology
www.neonet.ch
webmaster@neonet.ch