Clinic

Sturge-Weber syndrome

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Abstract

Sturge–Weber syndrome (SWS) is a rare congenital disorder belongs to a group of disorders collectively known as the phakomatoses (“mother-spot” diseases). It consists of congenital hamartomatous malformations that may affect the eye, skin, and central nervous system (CNS) at different times, characterized by the combination of venous angiomas of leptomeninges, face, jaws and oral soft tissues. We hereby report a 14 year old female presented with port wine stain and seizures and was diagnosed as Sturge-Weber syndrome after investigation (MRI). The co-occurrence of Sturge-Weber with facial nevus is 8% only.

Key words: cerebral atrophy, phakomatosis, port-wine stain

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Sturge-Weber syndrome (SWS) is believed to be caused by the persistence of vascular plexus around the cephalic portion of the neural tube. This plexus develops during the sixth week of intra-uterine development but normally undergoes regression during ninth week\(^1\). It is caused by a somatic activating mutation occurring in the GNAQ gene\(^2\). Phakomatoses are a group of neurocutaneous disorders characterised by involvement of structures that arise from the embryonic ectoderm (thus central nervous system, skin and eyes). Other organs may also be involved. SWS was first described by Schirmer in 1860. A more specific description was given by Sturge in 1879\(^1\). The syndrome is named after William Allen Sturge and Frederick Parkes Weber\(^2\).

Case report

A 14 year old female, known to using carbamazapine since the age of seven for a seizure disorder is presented with chief complaints of left-sided focal seizures. The seizures were clonic, involving her left arm, of 1 minute duration and followed by loss of consciousness for about 5 minutes. She later regained consciousness without any history of post-ictal confusion or residual weakness. She had missed her night dose of carbamazapine on the previous day. The seizures are not associated with any aura, headache, tinnitus, vomiting or fever. There was no history of head trauma, and no history of ear discharge with fever.

Birth history: The patient was born of a full term normal vaginal delivery at home; there was no history of birth trauma. Developmental milestones were normal. She was noticed to have facial pigmentation since birth. She had been fully vaccinated as per the prevailing vaccination schedule. There was however a history of surgery to right eye at 6 months of age for buphthalmos. At the time of her presentation to us, she was studying in class 8, with average scholastic performance. Her vision was normal and there was no history of behavioral abnormalities. Menstrual history: The patient attained menarche at the age of 13, and had regular menstrual cycles (4/30). Family history: She was second in birth order and no other family members suffered with similar complaints.
On examination: The patient was a moderately built girl with a port wine stain on the right side of her face extending on to left cheek. There was a flamus nevus on her left shoulder extending onto left upper arm and anterior chest. Her vital data were normal. A thorough examination of her central nervous system revealed no abnormality.

Investigations

Her routine investigations were normal, her EEG showed a normal awake study.

Discussion

Sturge–Weber syndrome (SWS) is a rare congenital disorder belongs to a group of disorders collectively known as the phakomatoses* (“mother-spot” diseases). Sturge-Weber syndrome (SWS), also called encephalo trigeminal angiomatosis, is a neurocutaneous disorder with angiomas that involve the leptomeninges (leptomeningeal angiomas and the skin of the face, typically in the ophthalmic (V1) and maxillary (V2) distributions of the trigeminal nerve. The hallmark of SWS is a facial cutaneous venous dilation, also referred to as a nevus flammeus or port-wine stain (PWS). It is observed at birth covering large part of face and cranium on one side. In one quarter of the cases nevus is bilateral. The nevus is deep red; its margins may be raised or flat. The involvement of upper eye lid is of great importance and nearly always associated with cerebral lesions. Meningial nevi are rarely the source of sub arachnoid cerebral hemorrhages. There can be seizures, low IQ, and underlying cerebral hemisphere atrophy as a result of chronic state of reduced perfusion and increased oxygen extraction. According to Nelson's Textbook of Pediatrics, the incidence of SWS is estimated at 1 person per 50,000. The inheritance is sporadic, with equal frequencies in boys and girls. No regional differences in incidence have been identified. SWS is referred to as complete when both CNS and facial angiomas are present and incomplete when only one area is affected without the other. The Roach scale is used for classification.
Type I - Both facial and leptomeningeal angiomas; may have glaucoma
Type II – Facial angiomas alone; may have glaucoma
Type III – Isolated leptomeningeal angiomas; usually no glaucoma.

Overall, about 50% of individuals with SWS will have glaucoma (optic neuropathy often associated with increased intraocular pressure), which can be present at birth or develop later. Increased pressure within the eye can cause the eyeball to enlarge and bulge out of its socket (buphthalmos)³.

Factors predicting a poor outcome (or indicating the need for surgery) in SWS include the following⁴: Increasing duration of postictal deficits, increasing focal or diffuse atrophy, progressive atrophy or calcifications, development of hemiparesis, deterioration in cognitive functioning (loss of intellectual abilities). Medical care in Sturge-Weber syndrome (SWS) includes anticonvulsants for seizure control, symptomatic and prophylactic therapy for headache, glaucoma treatment to reduce the intraocular pressure (IOP), and laser therapy for port-wine stain (PWS)². Latanoprost (Xalatan), a prostaglandin, may significantly reduce IOP (intraocular pressure) in patients with glaucoma associated with Sturge–Weber syndrome. Medical treatment of glaucoma in SWS usually fails with time, so most ophthalmologists consider surgical therapy to be the mainstay of treatment for SWS-associated glaucoma⁶. Surgery is desirable in patients with SWS for refractory seizures, glaucoma, and specific problems related to various SWS-associated disorders, such as scoliosis⁵. Surgical procedures include focal cortical resection, hemispherectomy, corpus callosotomy, and vagal nerve stimulation (VNS)⁶. SWS is considered one of the catastrophic epilepsies, which, according to Holmes⁷, result in poor seizure control and developmental outcome if not controlled early. However, criteria for medical intractability should be fulfilled before considering surgery. The PWS needs to be evaluated within the first week of life and differentiated from hemangioma. Treatment of the cutaneous PWS with dye laser photoacogulation has been helpful in reducing the cosmetic blemish from the cutaneous vascular dilatation⁸. The therapy should start as soon as possible, since multiple treatments are needed and earlier treatment may reduce the number of sessions required.

Clinical differential diagnosis of SWS includes Klippel-Trenaunay-Weber syndrome (port-wine stains of the extremities and face, hemihypertrophy of soft and bony tissues, with all of the characteristics of SWS), and Beckwith-Wiedemann syndrome (facial port-wine stain (PWS), macroGLOSSIA, omphalocele, and visceral hyperplasia. Several conditions may radiologically mimic SWS, e.g. the Dyke-Davidoff-Masson syndrome (one cerebral hemisphere is partially or completely atrophic as a result of an intrauterine or perinatal carotid artery infarction), severe siderosis (the typical contrast enhancement and the abnormal veins seen with contrast injection easily distinguish it from SWS), intrathecal methotrexate therapy related calcification and meningitis (neither of these demonstrate the unilateral specific geographic localization)⁹.

This patient had both facial and leptomeningial angiomas with cerebral calcification and therefore falls under Type I (Roach scale) and complete Sturge Weber syndrome. Since her seizure was precipitated on account of non-compliance with anti-epileptic medication, she was advised regular use of carbamazine and periodic follow up.


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References