Sturge–Weber syndrome

Key concepts

● Cardinal signs:

1) localized cortical cerebral atrophy and calcifications.

2) ipsilateral port-wine facial nevus (usually in distribution of V1)

● contralateral seizures usually present

● plain skull films classically show “tram-tracking” (double parallel lines).

AKA encephalotrigeminal angiomatosis. Sturge-Weber syndrome (SWS) is a rare, congenital neurocutaneous disorder involving the brain, skin & eye, consisting of:

1. cardinal features:

a) localized cerebral cortical atrophy and calcifications(especially cortical layers 2 and 3,with a predilection for the occipital lobes):

● calcifications appear as curvilinear double parallel lines (“tram-tracking”) on plain X-rays

● cortical atrophy usually causes contralateral hemiparesis, hemiatrophy, and homonymous hemianopia (with occipital lobe involvement)

b) ipsilateral port-wine facial nevus(nevus flammeus)usually in distribution of 1st division of trigeminal nerve (forehead and/or eyelid) (rarely bilateral): not always present, alternatively sometimes in V2 or V3 regions

2. other findings that may be present:

a) ipsilateral exophthalmos and/or glaucoma, coloboma of the iris

b) oculomeningeal capillary hemangioma

c) cerebral venous malformation (leptomeningeal angiomatosis)

d) convulsive seizures:contralateral to the facial nevus and cortical atrophy.Present in most patients starting in infancy

e) retinal angiomas

f) endocrinopathies: growth hormone deficiency is more common in SWS patients. For suspected or confirmed SWS, screen for this in children ≥ age 2 years by measuring serum IGF-1

Classification

Sturge Weber Syndrome can be classified into three different types. Type 1 includes facial and leptomeningeal angiomas as well as the possibility of glaucoma or choroidal lesions. Normally, only one side of the brain is affected. This type is the most common. Type 2 involvement includes a facial angioma (port wine stain) with a possibility of glaucoma developing. There is not any evidence of brain involvement. Symptoms can show at anytime beyond the initial diagnosis of the facial angioma. The symptoms can include glaucoma, cerebral blood flow abnormalities and headaches. More research is needed on this type of Sturge Weber Syndrome. Type 3 has leptomeningeal angioma involvement exclusively. The facial angioma is absent and glaucoma rarely occurs. This type is only diagnosed via brain scan.

Diagnostic criteria

Diagnosis is made based on having 2 out of 3 of the following:

● facial port-wine birthmark

● increased intraocular pressure

● leptomeningeal angiomatosis

Patients with only leptomeningeal angiomatosis and no skin or eye involvement are considered to have the intracranial varient of SWS.

▶ Other clinical aspects. Only 8–20% of patients with facial port-wine birthmarks (with or without ocular involvement) develop neurologic symptoms. Those with port-wine stain only in V2 and V3 have a lower risk of developing symptoms of SWS, and those with bilateral V1 birthmarks have a higher risk (≈ 35%).

Genetics

Most cases are sporadic, caused by a somatic mutation in a nucleotide transition in the gene GNAQ on chromosome 9q21 that occurs early after conception during in utero development.

Other cases are suggestive of recessive inheritance, with chromosome 3 being implicated.

Clinical features

Sturge–Weber syndrome clinical features.

Diagnosis

Sometimes associated with neurocutaneous melanosis.

Radiological findings will show tram track calcifications on CT, bilaterally.

Higher estimated myelin was observed on the ipsilateral side in some patients aged ≤ 2 years and lower myelin on the ipsilateral side in all older patients. Synthetic qMRI might be useful for showing myelin-related abnormalities in SWS [1)].

Treatment

Treatment is supportive. Anticonvulsants are used for seizures.

1. oxcarbazepine is a common initial drug. Side effects include central hypothyroidism, especially in girls

2. levetiracetam & topiramate are alternatives.

Skin lesions: laser treatment (currently, flashlamp-pumped PDL is favored) can lighten the birth-mark. It may also reduce hypertrophy of soft and bony tissue.

Endocrinopathies: growth hormone deficiency can be replaced; however, there may be a risk of increasing seizures.

XRT: complications are common and benefits are lacking.

Epilepsy Surgery in Sturge–Weber syndrome

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Case series

Sturge–Weber syndrome case series.

Case reports

An enhanced operative video presents the illustrative case of a total Posterior quadrant disconnection indicated for a 15-year-old boy with Sturge-Weber syndrome suffering from seizure recurrence after a partial PQD. Barrit et al. described the surgical procedure with emphasis on relevant anatomy and multimodal intraoperative guidance in three steps: (i) parieto-occipital disconnection, (ii) posterior callosotomy, and (iii) temporal disconnection/resection. Pearls and pitfalls of surgical management are discussed.

Posterior quadrant disconnection is a less invasive surgical option than typical hemispherectomy for selected indications of posterior multilobar epilepsy [2)].

A 2-year-old boy with Sturge-Weber syndrome who had in addition an intracranial lipoma, a temporal arachnoid cyst and a porencephalic cyst. This combination of intracranial lesions with Sturge-Weber syndrome has not been previously reported [3].

References

1)
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2)
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3)
Ergün R, Okten AI, Gezercan Y, Gezici AR. Sturge-Weber syndrome accompanied with multiple congenital intracranial lesions. Acta Neurochir (Wien). 2007 Aug;149(8):829-30; discussion 830. Epub 2007 Aug 1. PubMed PMID: 17660941.