Sturge-Weber syndrome

(Encephalotrigeminal Angiomatosis)

Last updated: April 17, 2019

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**Sturge-Weber syndrome** - congenital sporadic phacomatosis with capillary venous angiomas in ***leptomeninges***, ***skin of face***, ***eye***.

Pathophysiology

- ***residual embryonal blood vessels*** with secondary effects on surrounding brain tissue.

* + - normally, in 6th week vascular plexus develops around cephalic portion of neural tube, under ectoderm destined to become facial skin; vascular plexus regresses around 9th week of gestation.
    - failure of normal regression → angiomata.
    - neurologic dysfunction results from secondary effects:
      1. "vascular steal" around angioma → hypoxia, ischemia
      2. venous occlusion, thrombosis, infarction.
    - secondary effects are *aggravated by recurrent seizures* (even when short) → progressive dystrophic calcification\*, gliosis, atrophy → neurologic deterioration, seizures↑.

Although leptomeningeal angioma is static anatomic lesion, syndrome has progressive nature!

\*N.B. calcifications are located primarily in cerebral substance rather than in vessel walls

Etiology

- no recognizable genetic contribution; ***somatic mutations*** affecting:

* + - * 1. structure blood vessels (vessel circumference decreased, while vessel density increased)
        2. innervation of blood vessels (malformed vessels innervated only by noradrenergic sympathetic fibers)
        3. expression of extracellular matrix (fibronectin↓) and vasoactive molecules (endothelin-1 expression↑ in malformed vessels).

Epidemiology

incidence - 1 per 50,000

Pathology, Clinical Features

All lesions (if unilateral) tend to be ipsilateral!

No increased propensity for cancer!!!

1. **Leptomeningeal angiomas**
   * **unilateral** (85%) > bilateral (15%).
   * most common in *parietal* and *occipital* regions.
   * ipsilateral features - cerebral hemiatrophy, hemihypertrophy of skull and sinuses, enlarged choroid plexus, abnormal myelination.
   * cortical veins are either absent or replaced by few enlarged cortical veins.
   * neurologic manifestations:
2. seizures (72-93%) - typically focal; may be intractable; 75% before age of 1 year, 95% begin before 5 years.
3. **focal deficits** (esp. hemiparesis [25-56%], homonymous hemianopsia [44%]) - may be transient ("strokelike episodes"), but otherwise slowly progressive.
4. vascular headaches ("symptomatic migraine") (44-77%)
5. developmental disorders (50-75%) (developmental delay, learning disorders, mental retardation) - more common when angiomas are bilateral.

N.B. major intracranial hemorrhage is rare!

1. **Cutaneous angioma** (“port-wine stain” s. nevus flammeus) (87%) in skin of face.

N.B. most patients with facial port-wine stains do not have SWS!

N.B. presence of port-wine stain implies neither presence nor severity of intracranial leptomeningeal angiomatosis (only 8% of facial port-wine stains have this association)

* + typically in V1 and V2 distributions of CN5.

CNS is not affected if port-wine stain does not involve V1 area!

* + **unilateral** (49-86%; ipsilateral to CNS lesion) > bilateral (14-51%).
  + presents at birth – suspicion of diagnosis in neonate!
  + can be progressive (light pink macule → dark red or purple nodular lesion).

[Source of picture: H. Richard Winn “Youmans Neurological Surgery”, 6th ed. (2011); Saunders; ISBN-13: 978-1416053163 >>](http://www.amazon.com/gp/product/1416053166)

1. **Glaucoma** (30-71%) → buphthalmos (hydrophthalmia) → blindness.

N.B. glaucoma typically occurs only when port-wine stain involves eyelids!;

if port-wine stain is unilateral, glaucoma is ipsilateral!

* + causes: mechanical angle obstruction, episcleral venous pressure↑, secretion↑ of aqueous fluid (by choroidal hemangioma or ciliary body).
  + may be present at birth but can develop at any age.

1. **Eye hemangiomas** – choroidal (40%), conjunctival, episcleral.

Roach Scale classification

**Type I** - both *facial* and *leptomeningeal* angiomas; may have glaucoma.

**Type II** - *facial* angioma alone (no CNS involvement); may have glaucoma.

**Type III** - isolated *leptomeningeal* angiomas; usually no glaucoma.

Diagnosis

*Structural versus functional mismatch* (functional neuroimaging demonstrates greater area of involvement than structural neuroimaging) - especially important when considering epilepsy surgery!

**Skull x-ray** – pathognomonic subcortical *“tram-track” calcifications* in gyriform pattern (late finding – usually in patients > 2 yrs) - paired parallel lines that follow cerebral convolutions.

N.B. **calcification in ipsilateral\* outer cortex** rather than of blood vessels or white matter!

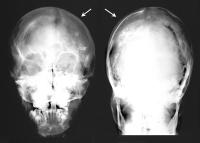
\*ipsilateral to port wine stain

* underlying ipsilateral cerebral atrophy → ipsilateral skull-table and orbital thickening, elevation of sphenoid wing and petrous ridge, enlarged ipsilateral paranasal sinuses and mastoid air cells.

“Tram-track” calcifications:

Smaller hemicranium on affected side:



**Angiography** – does not show angioma! (or early capillary blush)

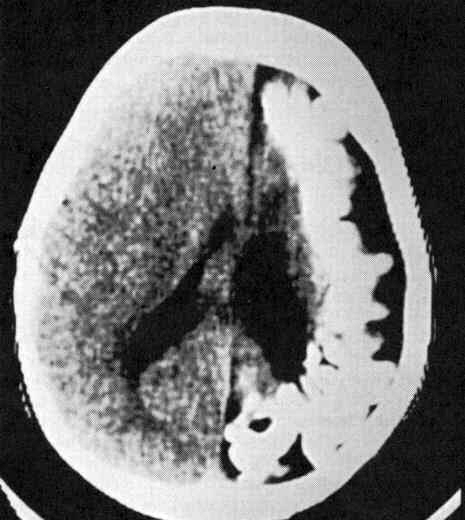
* lack of superficial cortical veins → nonfilling of dural sinuses (with absence of cortical veins, venous drainage occurs via enlarged tortuous transcortical veins into deep venous system).

**CT**:

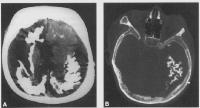
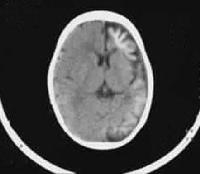
* *“tram-track” calcifications* under angioma (in infants and even neonates)
* adjacent cortical atrophy.

Shrunken cerebral lobe with calcified cortex

* enlarged ipsilateral choroid plexus & enlarged draining transcortical draining veins.
* BBB breakdown (during seizures).



CT - left hemiatrophy of cerebral cortex and typical gyral calcification:



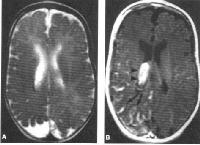
**MRI**:

* T2 - hyperintense leptomeningeal thickening and enhancement.
* gadolinium enhancement of angioma (appears as enhancement of subarachnoid space, medium covering cortical gyri and filling sulci) – early diagnosis!

N.B. enhancement is difficult to assess on CT in presence of calcification!

* adjacent **cortical atrophy**, accelerated / delayed myelination around angioma.
* enlarged ipsilateral choroid plexus (size correlates with angioma extent) & enlarged draining transcortical draining veins.
* progressive sinovenous occlusion → lack of superficial cortical veins (on MRV).

|  |  |
| --- | --- |
| Contrast T1-MRI - right cerebral atrophy, enhancing right occipital cortex, enlarged right choroid plexus:  Click to see larger picture | Contrast T1-MRI - intense pial enhancement and subjacent cerebral atrophy:  Click to see larger picture |



**SPECT**: hyperperfusion (in infancy, before first seizures) → classic *hypoperfusion* of involved hemisphere (after 1 year of age, even in those without epilepsy).

* steal phenomenon (during seizures).

**PET** – *hypometabolism*.

**EEG** – marked *voltage attenuation* in region of angioma; background suppression (74%); polymorphic delta activity; epileptiform discharges in remainder of cortex (22%).

**Biopsy** – typically not performed.

Treatment

* + 1. **Seizure control** improves neurologic outcome!

N.B. epilepsy surgery should not be delayed (ideally – during infancy)!

controversy regarding optimal timing of surgery: early surgery might preempt cognitive deficits from chronic, intractable epilepsy vs. early surgery might subject some patients to surgery risks.

* + - * complete seizure control is achieved in 10-50% patients; refractory seizures occur in 11-83%.
      * epileptogenic region is located in cortex adjacent to angioma.
      * localize ***area of seizure onset*** preoperatively by video EEG, ECOG, functional neuroimaging (e.g. ischemic regions may act as epileptogenic foci that may not be detected by CT / MRI).
        1. **Focal cortical resection** – when epileptogenic region is smaller and more localized.
        2. **Hemispherectomy** (anatomical hemispherectomy or functional hemispherectomy or hemidecortication) – for extensive, unilateral epileptogenic region; hemispherectomy is more successful if done during infancy!
        3. **Corpus callosotomy** – for bilateral disease (intractable atonic or tonic seizures leading to secondary injury).
        4. **Vagus nerve stimulation (VNS)** – for those who are not candidates for other surgical procedures.
    1. Prophylactic daily low-dose aspirin - for headaches, stroke-like events (may be result of progressive venous thrombosis).

N.B. varicella and yearly influenza immunizations (varicella / influenza + aspirin → Reye syndrome)

* + 1. Cosmetic ***laser therapy*** ASAP for port-wine stain (earlier treatment - fewer laser flashes needed to remove lesion).
    2. **IOP control**

Bibliography for ch. “Phakomatoses” → follow this [link >>](http://www.neurosurgeryresident.net/Pha.%20Phacomatoses,%20Neurocutaneous%20disorders\Pha.%20Bibliography.pdf)

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