

Sturge-Weber syndrome

(Encephalotrigeminal Angiomatosis)

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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:
 - a) "vascular steal" around angioma → hypoxia, ischemia
 - b) 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

- unclear; **somatic mutations** affecting:
 - a) structure blood vessels (vessel circumference decreased, while vessel density increased)
 - b) innervation of blood vessels (malformed vessels innervated only by noradrenergic sympathetic fibers)
 - c) 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!

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:
 - 1) **seizures** (72-93%) - typically focal; may be intractable; 95% begin **before 5 years**.
 - 2) **focal deficits** (esp. **hemiparesis** [25-56%], **homonymous hemianopsia** [44%]) - may be transient ("strokelike episodes"), but otherwise slowly progressive.
 - 3) vascular **headaches** ("symptomatic migraine") (44-77%)
 - 4) **developmental disorders** (50-75%) (developmental delay, learning disorders, mental retardation) - more common when angiomas are bilateral.

N.B. major intracranial hemorrhage is rare!

2. **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!

 - typically in V₁ and V₂ distributions of CN5.

CNS is not affected if **port-wine stain** does not involve V₁ area!
 - **unilateral** (49-86%) > bilateral (14-51%).
 - presents **at birth** – suspicion of diagnosis in neonate!
 - can be progressive (light pink macule → dark red or purple nodular lesion).



3. **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**.
4. **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

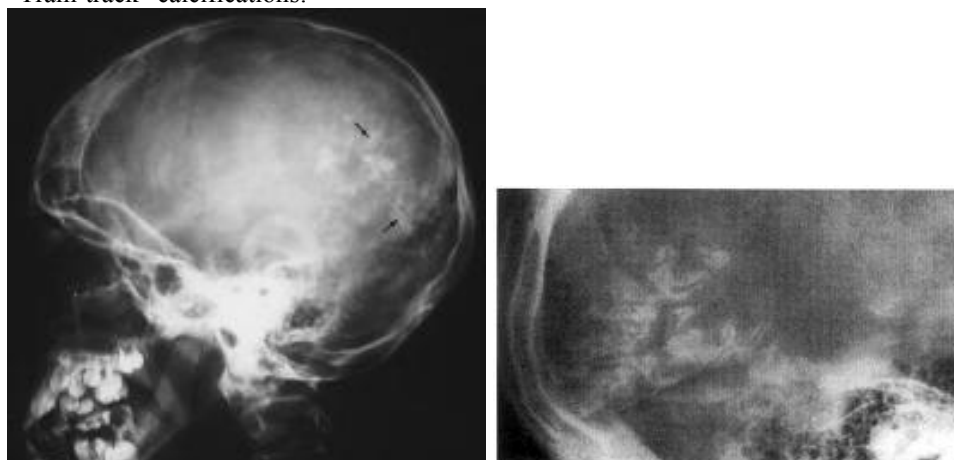
Structural versus functional mismatch (functional neuroimaging demonstrates greater area of involvement than structural neuroimaging) - especially important when considering epilepsy surgery!

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 outer cortex rather than of blood vessels!

- 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:

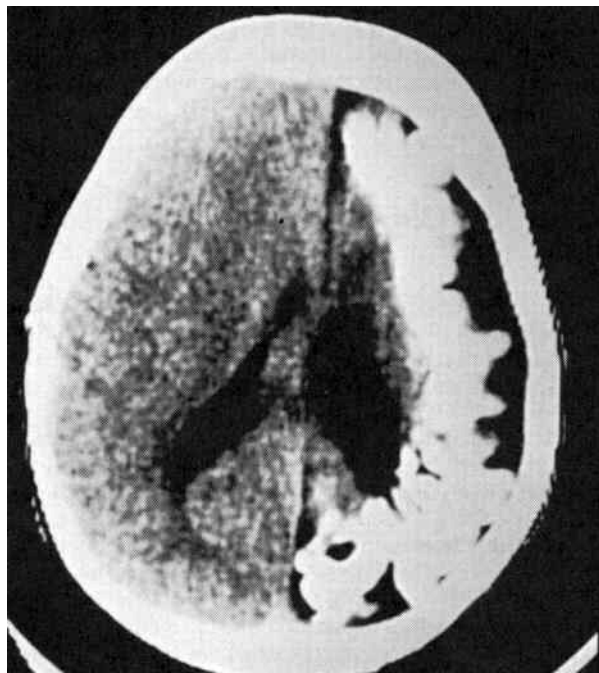


Angiography – does not show angioma! (or early capillary blush)

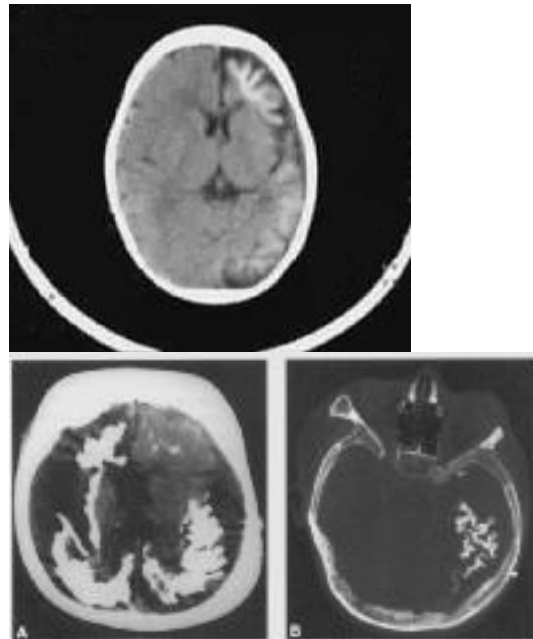
- lack of superficial cortical veins → nonfilling of dural sinuses (with absence of cortical veins, venous drainage occurs via enlarged tortuous transcortical veins into deep venous system).

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).



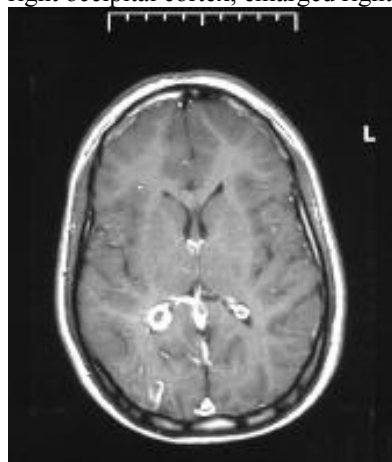
CT - left hemiatrophy of cerebral cortex and typical gyral calcification:



MRI:

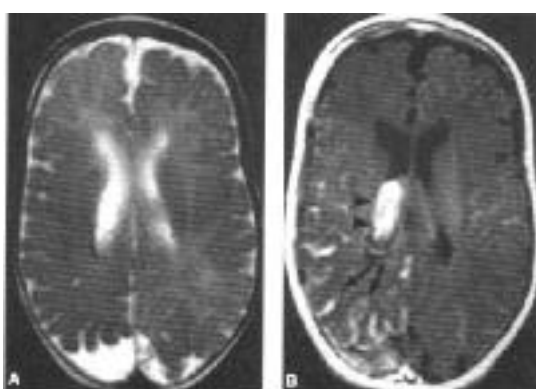
- T2 - hyperintense leptomeningeal thickening.
- gadolinium enhancement of angioma** (appears as enhancement of subarachnoid space, medium covering cortical gyri and filling sulci) – early diagnosis!
 - N.B. this enhancement is difficult to assess on CT in presence of calcification!
- adjacent cortical atrophy, accelerated / delayed myelination around angioma.
- enlarged ipsilateral choroid plexus (size correlates with angioma extent) & enlarged draining transcortical draining veins.
- progressive sinovenous occlusion (on MRV).

Contrast T1-MRI - right cerebral atrophy, enhancing right occipital cortex, enlarged right choroid plexus:



Contrast T1-MRI - intense pial enhancement and subjacent cerebral atrophy:





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)!

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

BIBLIOGRAPHY for ch. “Phakomatoses” → follow this [LINK >>](#)