The Roles of Radiologic Imaging in Sturge-Weber Syndrome

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Overview

• Patient TR
• The Phakomatoses
• Sturge-Weber Syndrome
  – Clinical Features
  – Epidemiology
  – Pathogenesis
• Patient TR: Imaging
• Companion Patient RW
• Summary
Patient TR

- Newborn baby girl with a L-sided Port-Wine Stain primarily in the V1 distribution at birth
- L-sided ocular glaucoma

What should we be concerned about?

http://www.childrenshospital.org/clinicalservices/Site2566/mainpageS2566P7.html
Patient TR: Sturge-Weber Syndrome?

She has classic features of Sturge-Weber Syndrome, a phakomatosis.
The Phakomatoses

• “Phakos” (Gr.) = birth mark, spot, mole
• Neurocutaneous syndromes/Congenital neuroectodermal dysplasias:
  – Neurofibromatosis
  – Tuberous Sclerosis
  – Von Hippel Lindau
  – Sturge-Weber Syndrome (SWS)
    • the only phakomatosis that is NOT associated with intracranial neoplasms (*Di Rocco and Tamburrini, 2006*)
SWS: Classic Clinical Features

- Encephalotrigeminal angiomatosis:
  - Capillary-venous malformation (leptomeningeal angiomatosis)
  - Facial port-wine stain (PWS or nevus flammeus) in trigeminal V1-V3 distribution
  - Congenital glaucoma
  - Intractable epilepsy
  - Progressive mental retardation

http://www.childrenshospital.org/clinicalservices/Site2566/mainpageS2566P7.html
SWS: Epidemiology

- Rare (estimated 1/50,000 live births)
- Sporadic
- Affects males and females with equal frequency
- No racial bias

(Di Rocco and Tamburrini, 2006)
SWS: Classification

<table>
<thead>
<tr>
<th>SWS Type</th>
<th>Clinical Features</th>
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<tbody>
<tr>
<td>1 (“classic”)</td>
<td>Facial and intracranial manifestations</td>
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<tr>
<td>2</td>
<td>Facial lesion only (primarily dermatological)</td>
</tr>
<tr>
<td>3</td>
<td>Intracranial manifestations \textit{without} facial lesions</td>
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(Adapted from Tortori-Donati et al, 2005)

• Overall risk of PWS associated with leptomeningeal angiomatosis: ~8%
  – In the first year of life, 75-90% of these patients develop seizures, about 60% of which become progressively refractory to medical treatment
  – May require surgical lobectomy or hemispherectomy if severe

(Di Rocco and Tamburrini, 2006)
SWS: Pathogenesis

• SWS is thought to develop from localized primary venous dysplasia, unrelated to any trigeminal nerve dysfunction.
  – The facial distribution of PWS appears to be coincidental (Parsa, 2008).

• During development at ~4-5 weeks gestation, a primordial sinusoidal vascular plexus forms around the cephalic portion of the neural tube and under the ectoderm that later becomes facial skin.
  – This vascular plexus normally regresses at ~9 weeks gestation (Tortori-Donati et al, 2005).
SWS: Pathogenesis

- In SWS, cortical bridging veins fail to form, the vascular plexus persists, and the remaining veins become engorged with redirected blood flow (Parsa, 2008).

Intraoperative images showing leptomeningeal “angiomatosis,” reflecting venous engorgement with diffuse hemispheric involvement in (a) and more focal involvement in (b). (Di Rocco and Tamburrini, 2006)
Cerebral Veins are Emissary Veins

* **Key point:** Because all emissary veins lack valves, they allow for **bidirectional** flow.

Let’s review the **cerebral venous system** to better understand the key features of SWS.
The superficial parts of the brain are primarily drained by the superior sagittal sinus, while the deeper structures are drained by the cavernous sinus, straight sinus, and other deep veins. The cortical bridging veins serve as the conduits that connect the two venous systems, which ultimately drain into the internal jugular vein.
Abnormal Venous Drainage in SWS

- With absent or fewer cortical bridging veins, normal cerebral outflow is impaired, leading to engorgement of the remaining veins.
- Since cerebral veins have bidirectional flow, venous blood can travel to superficial (e.g. leptomeningeal, facial) veins or to deep veins/sinuses, including the choroidal plexus veins and ophthalmic veins.

(Modified from Parsa, 2008)
Neurologic Deterioration in SWS

• Processes (including normal brain development and seizures) that increase the oxygen and glucose demand of brain tissue lead to increased cerebral blood flow.
  • These changes exacerbate the pre-existing venous engorgement and further elevate venous pressures, resulting in more severe cerebral ischemia and tissue damage (Parsa, 2008).

• With this in mind, let’s return to Patient TR…
Back to Patient TR

• Newborn baby girl with a L-sided Port-Wine Stain in primarily the V1 distribution at birth

• L-sided ocular glaucoma

• We are concerned about intracranial involvement of SWS.

Which imaging modality should we use?
Pt TR: MRI Brain (4 days old)

- MRI is optimal for soft tissue imaging; therefore, it is the modality of choice to evaluate for white or gray matter alterations, vascular abnormalities, and parenchymal volume loss.
  - Superior to CT for correlation with clinical status (Marti-Bonmati et al, 1993)
- Axial T1-weighted FLAIR, post-gadolinium:
  - Slight asymmetry of the vessels over the L high convexity; no other significant findings

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Pt TR: MRI Brain (4 days old)

• History of L-sided glaucoma and choroid “hemangioma”

Increased enhancement, thickness of L choroid relative to R

Not a true hemangioma; instead, reflects engorgement of pre-existing vessels, related to increased ocular venous pressure (Parsa, 2008)

Axial T1-weighted post-gad

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Pt TR: At 8 Months of Age

- Patient TR presented to the ED with new-onset seizures/status epilepticus, R facial droop, and little spontaneous movement of her R arm.
- No head trauma
- No family history of seizures
- Afebrile, vital signs within normal limits

What kind of imaging would you do to R/O possible causes of her seizures?
Pt TR: CT Brain (8 months of age)

- R/O acute intracranial hemorrhage/infarction as the most urgent possibilities, also evaluate for mass lesions and hydrocephalus

- Non-contrast Axial CT:
  - No sign of acute bleed, territorial infarct, mass lesions, or hydrocephalus
  - Bilateral **subcortical mineralization** involving the parietal and temporal lobes (L>R)
  - Parenchymal volume loss (L>R)
DDx of Intracranial Calcifications

- Sturge-Weber syndrome
- Arteriovenous malformation
- Hemangiomas
- Choroid plexus
- Craniopharyngioma
- Glioma
- Tuberculosis
- Idiopathic

(Reeder, 2003)
“Tram-tracking” in SWS

- Gyriform cortical calcifications on CT and plain film due to:
  - Altered vessel wall permeability → leakage of calcium phosphate or carbonate with subsequent secondary crystallization within perivascular parenchyma
  - Dystrophic calcification: related to prolonged convulsive states with local ischemia → anoxia, neuronal necrosis

- Calcifications develop over time, usually beginning at >2 yrs of age

(Posteroanterior skull radiograph. (Akpinar, 2004)

(Di Rocco and Tamburrini, 2006)
Pt TR: MRI Brain (8 months of age)

- Evaluate for soft tissue changes, vascular abnormalities
- Axial MPGR Susceptibility:
  - Progressive cerebral atrophy with widened subarachnoid space
  - Increased leptomeningeal enhancement involving both cerebral hemispheres (L>R)

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Pt TR: At 2 Years of Age

- Patient TR presented to the ED with an increasing frequency of seizures and a change from her baseline seizure characteristics

- Developmental delay was noted on exam

What kind of imaging would you do?
PT TR: CT Brain (2 yrs of age)

- R/O intracranial bleed due to concern for change in seizure characteristics; evaluate for SWS progression

- Non-contrast Axial CT:
  - Interval progression of marked gyriform cerebral calcification and associated parenchymal volume loss (L>R).

Compare with prior CT (at 8 mo):

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Pt TR: MRI Brain (2 yrs of age)

- Evaluate for interval progression of SWS features
- Axial T1-weighted, post-gad:
  - More pronounced b/l leptomeningeal enhancement, especially within R parietal, temporal, occipital lobes
  - Engorgement of choroid plexus (related to impaired venous outflow)
  - Choroid plexus cysts
Pt TR: MRI Brain (2 yrs of age)

- Axial gradient echo:
  - Dilated collateral veins
  - Calcifications appear dark

Calcifications are more prominent on CT:

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Patient TR: Management

- Patient TR’s seizures responded to oxcarbazepine and valproic acid.

- She was discharged home on these anticonvulsants with instructions to follow up with the neurology service.
Companion Patient RW: History

• Boy with Sturge-Weber Syndrome with associated R-sided facial port-wine stain in V1 distribution
• Diagnosed at 5 weeks of age with SWS based on MRI findings
• First seizures presented at 6 months of age

Let’s look at some imaging.
Pt RW: What do you see?

Leptomeningeal enhancement

Contrast-enhanced Axial T1 weighted MRI (5 wks of age)

Cortical calcifications

Non-contrast Axial CT (6 months of age)

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Pt RW: Rapid Progression on MR
(Axial T1-weighted Post-gad Images)

Leptomeningeal enhancement

Parenchymal atrophy

Choroid plexus enlargement

5 wks of age

9 mo of age

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Pt RW: Progression to Intractable Seizures

• Patient RW continued to have seizures, which became refractory to medical therapy.

• Surgical options were considered to remove the epileptogenic focus and protect the normal brain from excitotoxic injury secondary to the seizures.

How can we functionally determine the extent of brain involvement?
Nuclear Imaging: PET

• Positron Emission Tomography

• F-18-FDG (Fluorodeoxyglucose)
  – Measure F-18-FDG metabolism in brain tissue to determine its metabolic function

• More recently used for prognostic evaluation and to aid surgical planning
  – Extent and severity of FDG hypometabolism have been shown to correlate with seizure severity and cognitive decline (Lee et al, 2001)
Pt RW: Pre-surgical Evaluation with Brain PET

PET scan shows decreased FDG uptake throughout the R cerebrum, suggesting decreased metabolic activity within the entire hemisphere, which correlates with the region of cerebral hemiatrophy seen on MRI.

Normal FDG uptake is denoted by the yellow regions.
Pt RW: Management

PET scanning demonstrated diffuse reduction in metabolic activity throughout the entire R cerebrum.

Therefore, the decision was made for Patient RW to undergo R cerebral hemispherectomy.
Pt RW: s/p R Cerebral Hemispherectomy

Contrast-enhanced Coronal T1w MRI

Contrast-enhanced Axial T2w MRI

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Let’s Review

• You have been introduced to two different patients with Sturge-Weber Syndrome.
• Both demonstrated classic radiologic findings on MRI and CT but clinically progressed at different rates.
• One patient’s seizures were managed medically, but the other patient’s became intractable, requiring surgical intervention.

We can use what we have learned from these two patients to construct a simple algorithm for the diagnosis and management of SWS patients in general.
SWS: Algorithm for Dx and Management

Facial port-wine stain at birth in V1-V3 distribution

R/O intracranial involvement with radiologic imaging

Leptomeningeal enhancement and parenchymal atrophy (MRI); gyriform calcifications (CT)?

No

Repeat imaging if develop clinical symptoms (e.g. seizures)

Yes

Close neurology follow-up with radiologic documentation of intracranial progression if clinical status changes; manage seizures medically

Consider surgical management if refractory to medical therapy; employ functional imaging (PET) for prognostic and pre-surgical evaluation

Dermatologic evaluation for laser Rx of nevus for cosmetic concerns

Ophthalmologic evaluation for glaucoma
Summary

• **Sturge-Weber Syndrome** is a rare, sporadic condition due to primary venous dysplasia causing impaired venous outflow and subsequent cerebral ischemia.

• Certain types can progress to intractable epilepsy and may necessitate radical surgical intervention.

• Radiologic imaging plays several key roles in the management of SWS patients.
Summary - 2

• MRI correlates better than CT with clinical progression.
  – Confirms the diagnosis of intracranial involvement
  – Helps document the extent of involvement (e.g. R/O bilateral SWS)

• CT is more sensitive than MRI for detecting cortical calcifications.
Summary - 3

• PET offers functional (metabolic activity) data to determine the full extent of involvement of the brain parenchyma.
  – Complements MRI/CT findings
  – Provides prognostic information that can help guide management, including surgical planning
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