# Convulsión en niño con angioma facial

Alfredo Jordán García

R1

Sección Neurología Pediátrica: Francisco Gómez Gosálvez y Rocío Jadraque Rodríguez

26/11/2014

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Enfermedad actual

- Lactante de 11 meses
- Febrícula. Otitis
- Convulsión tónico-clónica hemicuerpo izquierdo
- Desviación de mirada hacia la izquierda
- TAC: hemorragia subaracnoidea en lóbulo parietal derecho
- Analítica sanguínea normal

Servicio de Pediatría
DEPARTAMENTO DE SALUD ALICANTE - HOSPITAL GENERAL

# Antecedentes personales y familiares

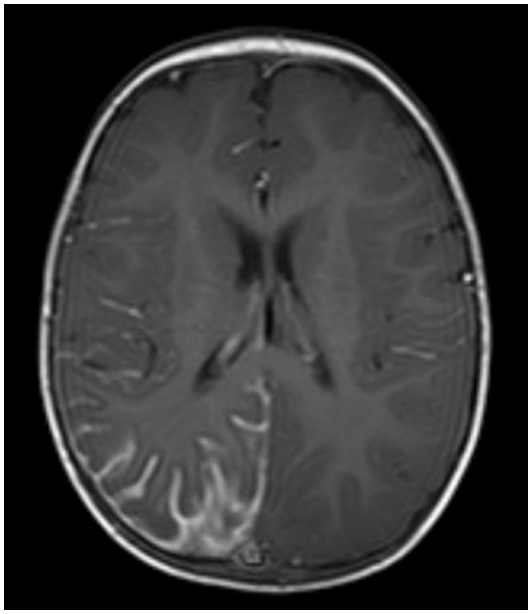
- Embarazo normoevolutivo y controlado
- Parto vaginal. EG 39+4
- Perinatal sin incidencias
- Cribados metabólico y auditivo normales
- Angioma plano vinoso en 1ª rama trigémino derecho
- ECO (enero/2014)
- RMN (junio/2014)

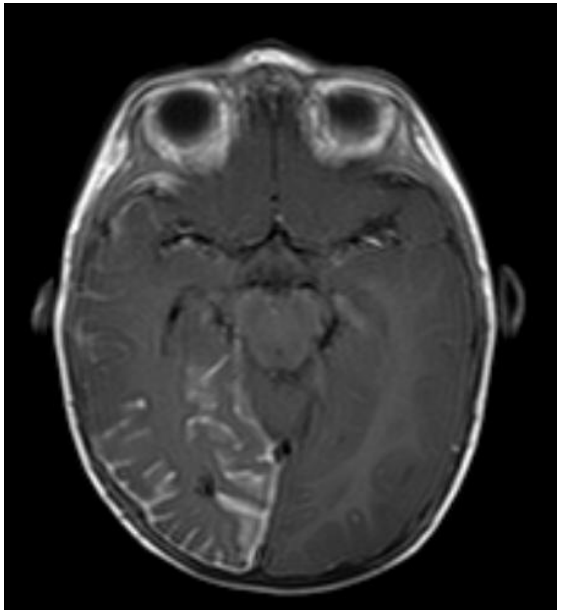
Servicio de Pediatría
DEPARTAMENTO DE SALUD ALICANTE - HOSPITAL GENERAL

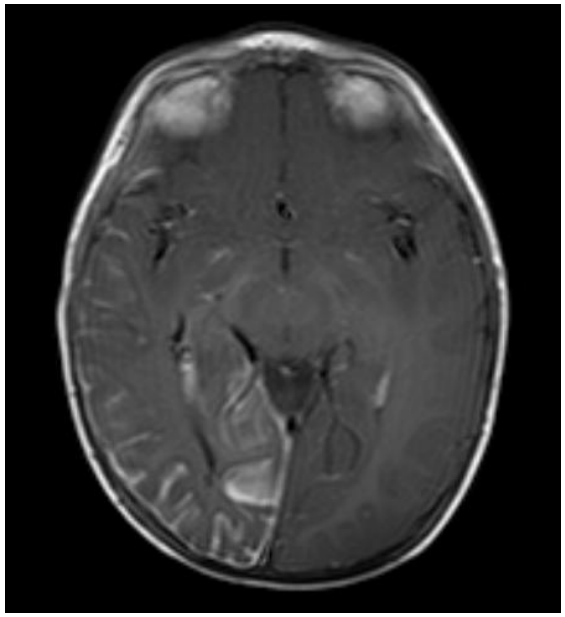
# Exploración física

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

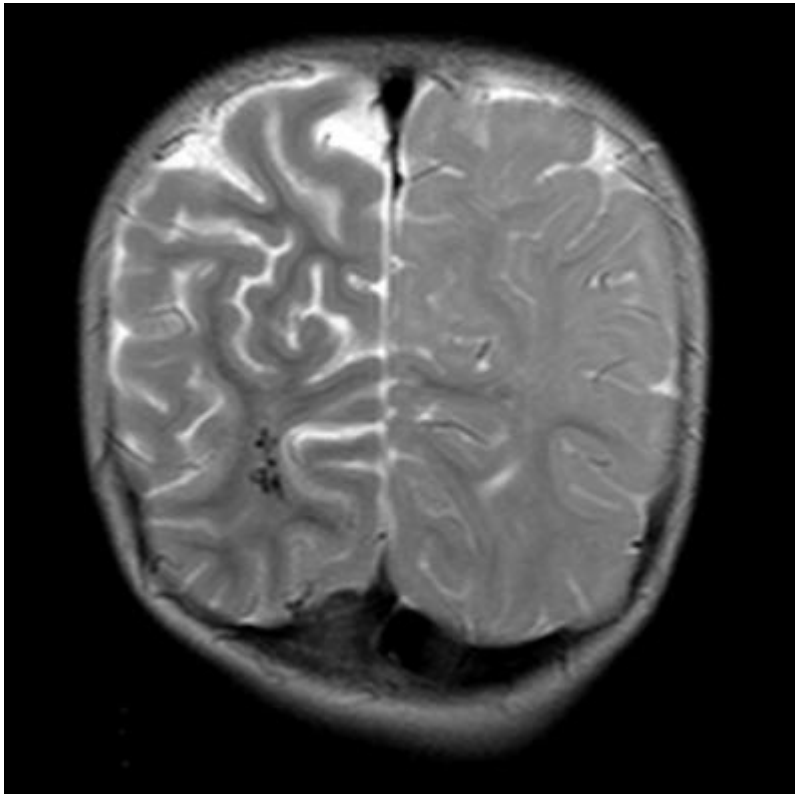
# Pruebas complementarias

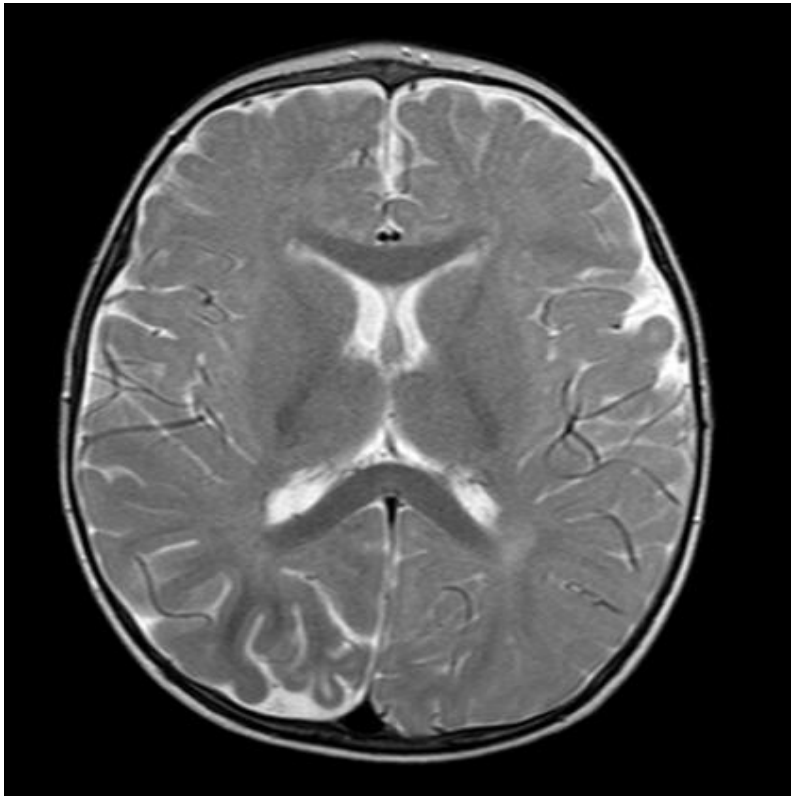






Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL





Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Evolución

- Convulsión parcial compleja izquierda cede con diazepam
- Tratamiento de mantenimiento: Keppra (levetiracetam)


Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Síndrome Sturge-Weber (SWS)

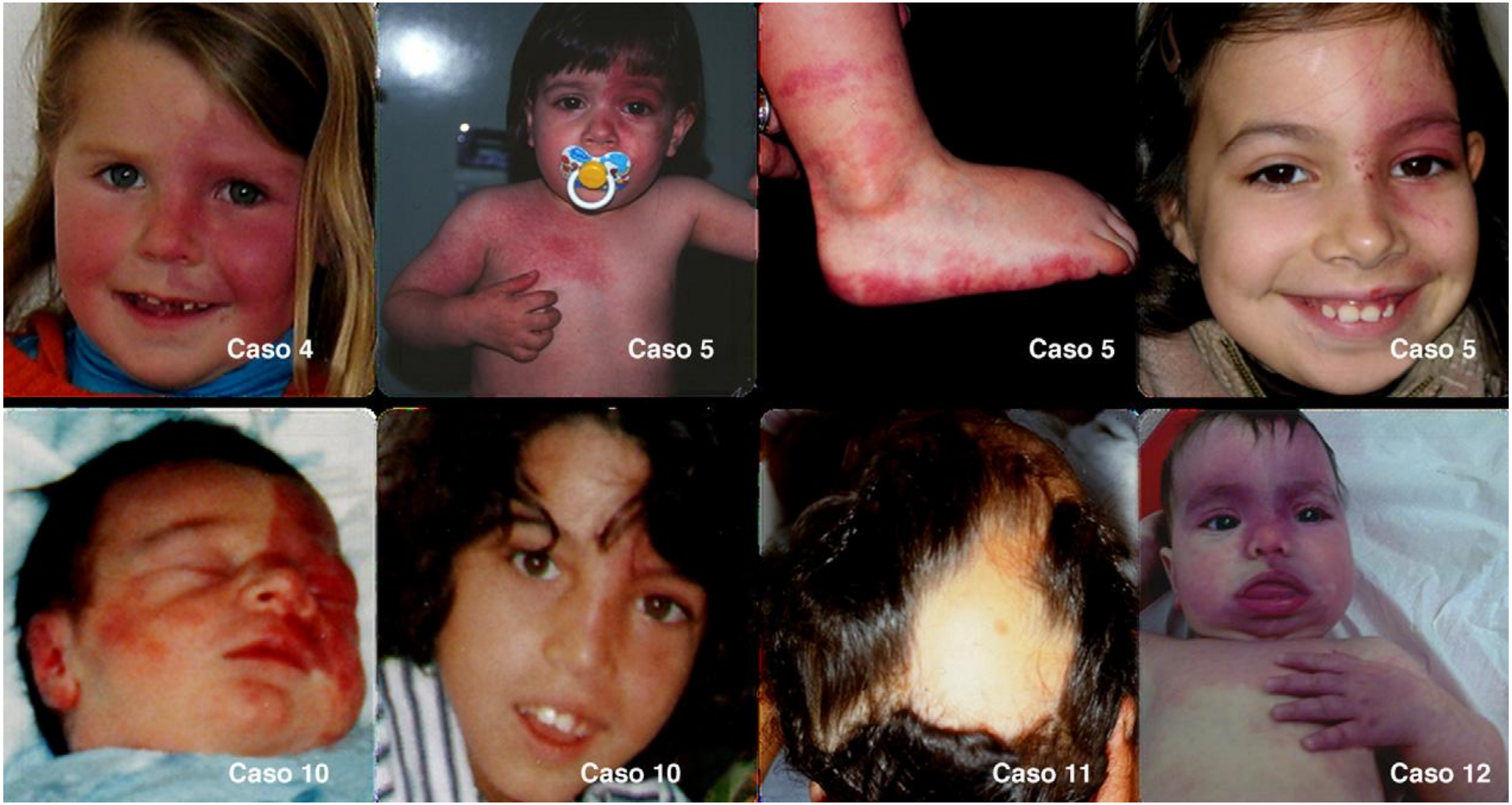
# Genética y Patogenia

- Mutaciones gen GNAQ
- Ectodermo
- Mutación precoz: SWS
- Mutación tardía: no síndrome.

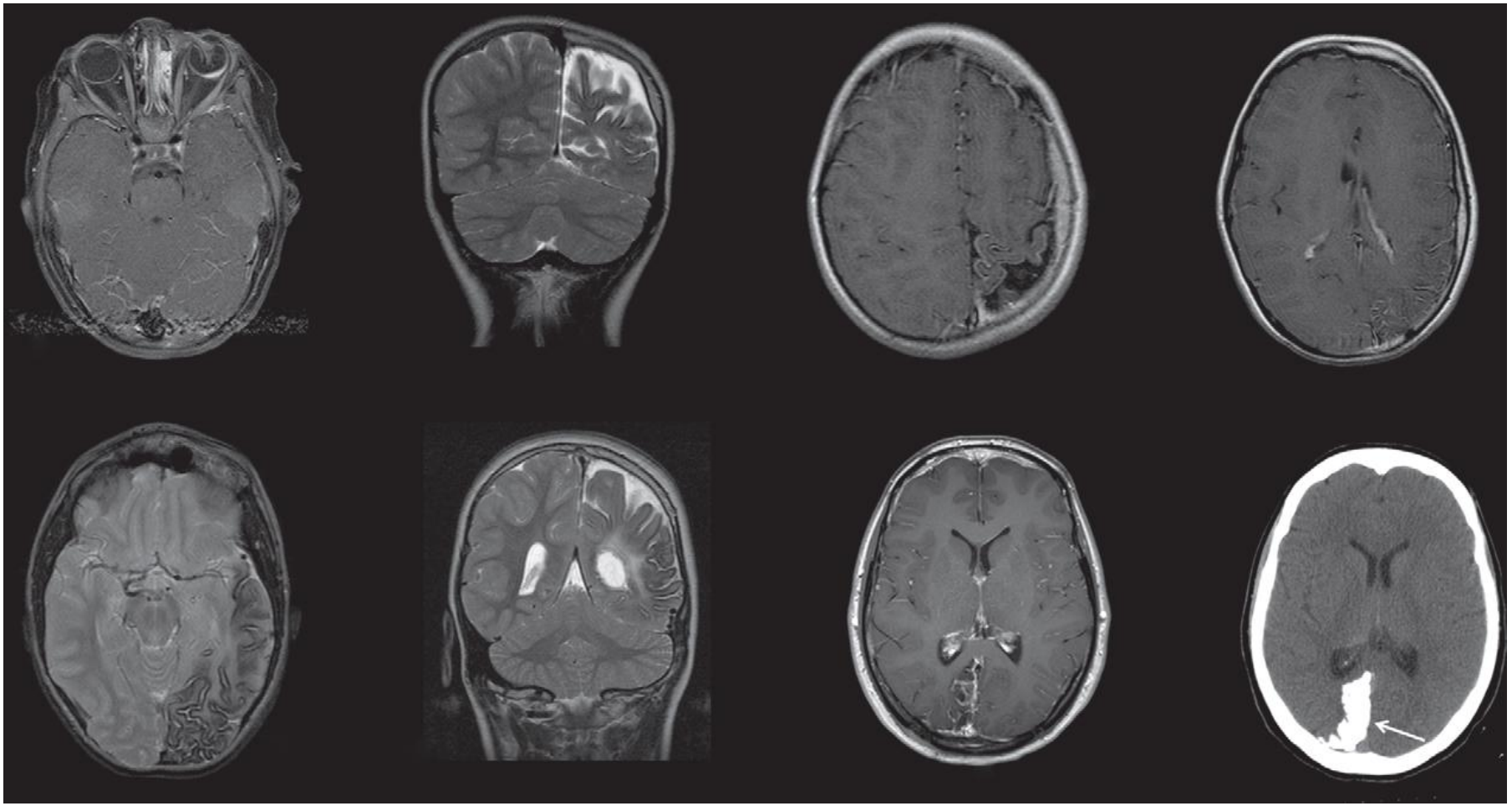
Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Manifestaciones Clínicas

- Angioma rojo vinoso
- Malformación leptomeníngea
- Convulsiones
- Hemiparesia
- Retraso mental
- Problemas de comportamiento
- Defectos visuales
- Glaucoma
- Otras manifestaciones oftalmológicas
- Aspectos neuroendocrinos
- Comorbilidades




Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL



Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

ELSEVIER

# Autism with Facial Port-Wine Stain: A New Syndrome?

Harry T. Chugani, MD*†‡, Csaba Juhász, MD, PhD*†, Michael E. Behen, PhD*†, Ross Ondersma, MD*†, and Otto Muzik, PhD*‡

**The hallmark of Sturge-Weber syndrome is leptomeningeal angiomatosis. Over 15 years, four children were identified (2 boys, age 2.9-6 years) with unilateral facial port-wine stain, referred for presumable Sturge-Weber syndrome but who were also autistic. Computed tomography and magnetic resonance imaging scans failed to show evidence of leptomeningeal angioma in all four children. Three of the children had a history of seizures. Detailed neuropsychologic testing of three children revealed a similar presentation, characterized by developmental disturbance, particularly involving delayed onset of language, and early-emerging social atypicality. Positron emission tomography scanning of cerebral glucose metabolism revealed hypometabolism in the bilateral medial temporal regions, anterior cingulate gyrus, frontal cortex, right temporal cortex, and cerebellum. The pattern of glucose hypometabolism differed from that of 12 children with infantile autism (age 2.7-7.9 years) who had mild left medial temporal but more severe right temporal cortical hypometabolism and showed a reversal of normal frontotemporal asymmetry of glucose metabolism. Unilateral facial port-wine stain and autism with no intracranial angioma on conventional imaging may represent a rare clinical entity distinct from both infantile autism and previously described variants of Sturge-Weber syndrome.** 

Chugani HT, Juhász C, Behen ME, Ondersma R, Muzik O. Autism with facial port-wine stain: a new syndrome? Pediatr Neurol 2007;37:192-199.

## Introduction

Sturge-Weber syndrome, or encephalofacial angiomatosis, is characterized by the presence of facial port-wine stain and leptomeningeal angiomatosis [1]. Port-wine stain is seen in 3 per 1000 newborns, but when this birthmark involves only the forehead and upper eyelid, leptomeningeal angioma is present in 10-20% of cases; the intracranial angioma is much less common when the lower face or trunk are affected by the port-wine stain. When present, the leptomeningeal venous angioma is typically unilateral, but it may be bilateral in 10-15% of patients. It may involve the entire hemisphere or a portion (usually the posterior aspect) of the hemisphere, leading eventually to cell death and calcification of the tissue underlying the intracranial angioma. The neurologic consequences of the angioma may include epilepsy, cognitive delay, and hemiparesis, hemiatrophy, and hemianopia contralateral to the side of the facial port-wine stain [2]. Glaucoma may be present in 30-70% of patients with Sturge-Weber syndrome.

Over 15 years, four children with facial port-wine stain and autism were evaluated at the Children's Hospital of Michigan, but with no evidence of leptomeningeal angiomatosis or calcifications on computed tomography and magnetic resonance imaging scans. Because these children appeared to be distinct from patients with Sturge-Weber syndrome and from those with infantile autism, classification for these children was unclear. The goal of the present study was to provide a detailed clinical description of the four children and to compare their positron emission tomography findings with those seen in normal subjects and in infantile autism.

Pediatric Neurology 48 (2013) 143–145

Contents lists available at ScienceDirect

Pediatric Neurology

journal homepage: www.elsevier.com/locate/pnu

Case Report

# Atypical Imaging Evolution of Sturge-Weber Syndrome Without Facial Nevus

**Rebecca R. Luke MS [a], Saleem I. Malik MD [b], Angel W. Hernandez MD [b], David J. Donahue MD [c], M. Scott Perry MD [b,*]**

[a] *University of North Texas Health Science Center, Fort Worth, Texas*
[b] *Comprehensive Epilepsy Program, Cook Children's Medical Center, Fort Worth, Texas*
[c] *Department of Neurosurgery, Cook Children's Medical Center, Fort Worth, Texas*

ARTICLE INFORMATION

*Article history:*
Received 30 August 2012
Accepted 4 October 2012

ABSTRACT

We report a patient with Sturge-Weber syndrome without facial angioma, who presented with seizures and normal initial imaging results. The patient experienced several years without seizures before a sudden increase in seizure frequency, followed by an atypical evolution of imaging findings prompting biopsy to establish the diagnosis. This case highlights not only the rare presentation of isolated leptomeningeal angiomatosis, but also the potential for atypical evolution of imaging findings through the course of the disease. We detail the imaging findings of our case and review the potential pathophysiological basis for this appearance. Our experience suggests that repeat imaging is warranted in patients with suspected Sturge-Weber syndrome or those with intractable cryptogenic epilepsy, because some imaging features of Sturge-Weber syndrome may manifest over time.

# Subtipos

- Tipo I (clásico): angioma facial + angioma leptomeníngeo +/- glaucoma
- Tipo II: angioma facial + glaucoma – angioma leptomeníngeo
- Tipo III (raro): angioma leptomeníngeo

# Diagnóstico

- Angioma facial + malformación leptomeníngea
- Distinguir del Síndrome Klippel-Trenaunay

- RMN con Gadolinio:

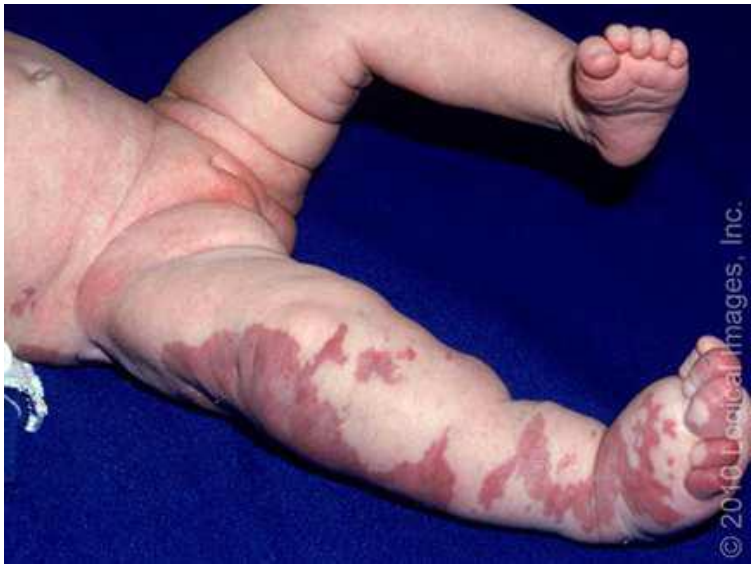
✓ Clínica neurológica

✓ >1 año sin clínica

- 90% pacientes con angioma sin lesión intracraneal

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Diagnóstico diferencial




Síndrome Klippel-Trenaunay

Síndrome PHACE

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Tratamiento

- ¿Aspirina?
- Fototermolisis con láser
- Tratamiento tópico de glaucoma o quirúrgico
- 40% controlado con anticonvulsionantes
- Refractarios:
  - ❑ Hemisferectomía

✓ 81% sin convulsiones

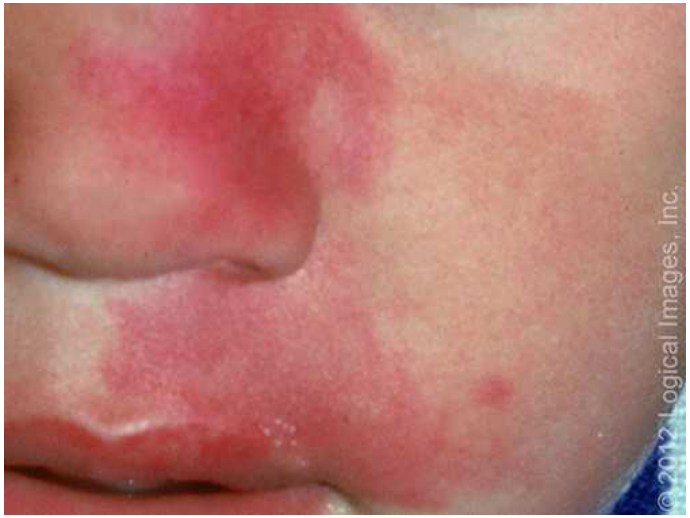
✓ 53% sin medicación

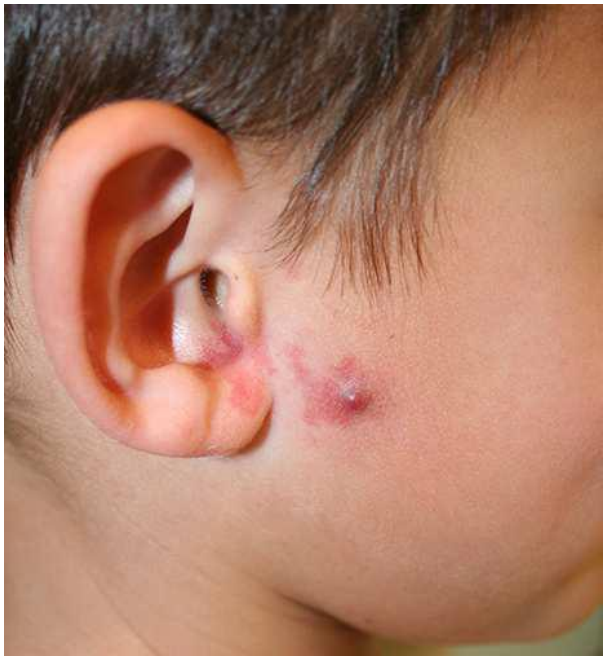
- ❑ Resección focal

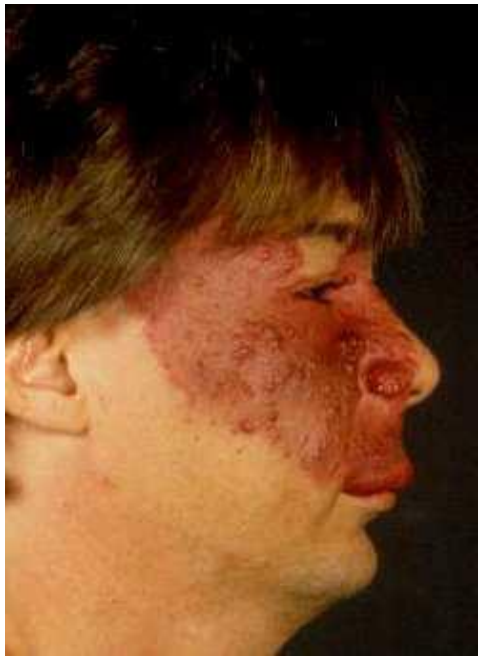
✓ 58% sin convulsiones

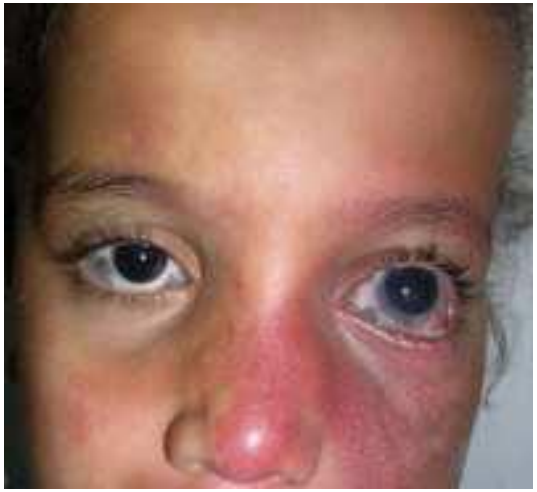
- ❑ Transección del cuerpo calloso
- ❑ Estimulación del Nervio Vagal

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL



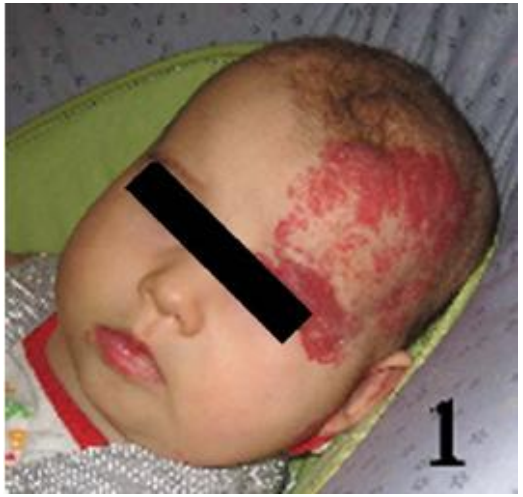



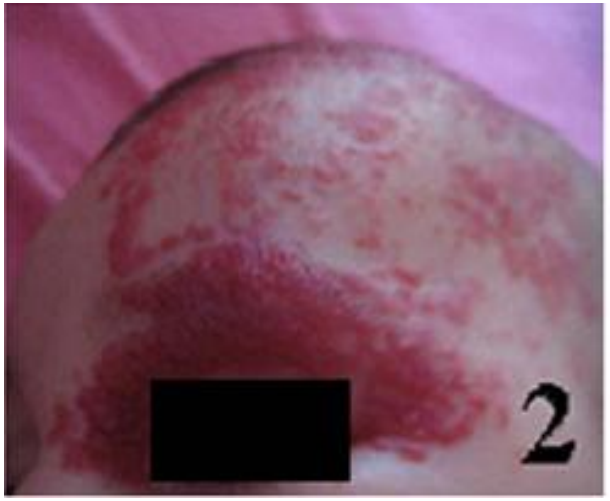


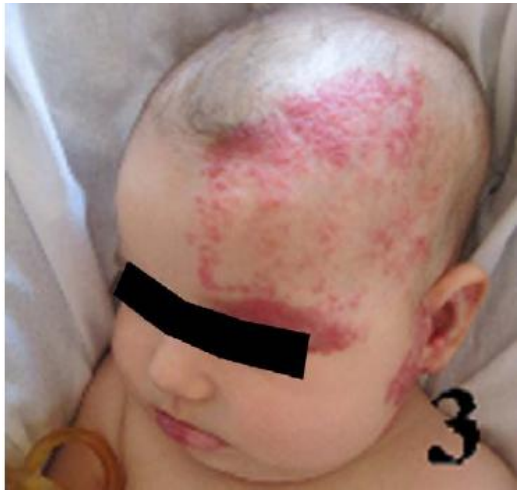


Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

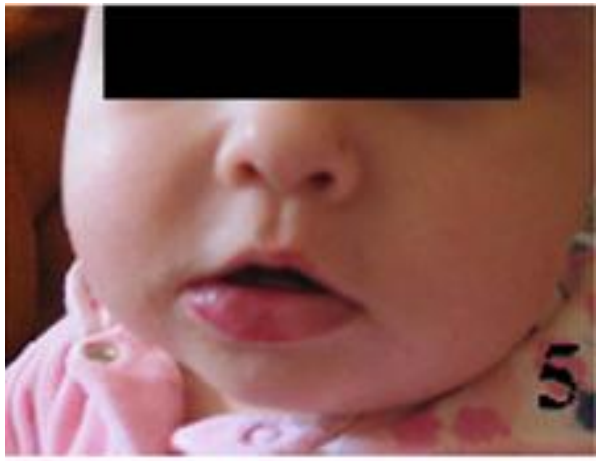
# Tratamiento del hemangioma infantil con Propranolol

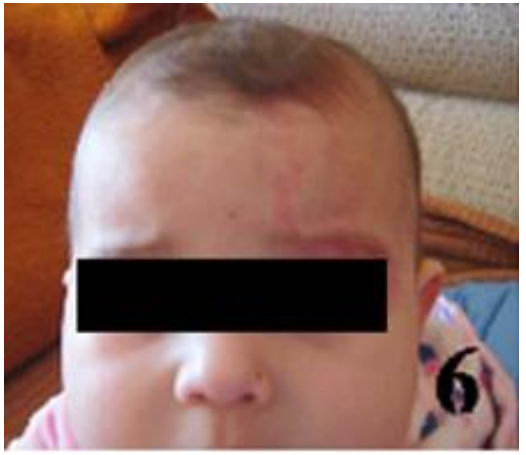






# ¿Cuándo operar?

- Epilepsia resistente a medicación

  *Pobre control de epilepsia tras 6 meses con un mínimo de 2 anticonvulsionantes*

Servicio de Pediatría
DEPARTAMENTO DE SALUD
ALICANTE - HOSPITAL GENERAL

# Pronóstico

- Extensión de malformación leptomeníngea
- Perfusión córtex cerebral
- Afección ocular
- Edad de comienzo epilepsia
- Respuesta a medicación