



Sturge Weber Syndrome/ Klippel Trenauay Syndrome

An overlap

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Case History

- ▶ New Born
- ▶ Normal delivery 3.1 kg
- ▶ Bilateral Port Wine Stain on face and several abnormalities
- ▶ Seizure

Examination & Investigations Findings

- ▶ Glucoma
- ▶ ASD/VSD
- ▶ Persistence of left SVC
- ▶ Focal seizures
- ▶ Cerebral atrophy
- ▶ Cerebral vascular malformation
- ▶ Laryngomalacia
- ▶ Hypochromic Anemia
- ▶ Limbs hypertrophy
- ▶ Facial port-wine stain
- ▶ Cutaneous lesions overall body (combined vascular malformation of capillaries, veins and lymphatics)

Haziness of
cornea

Larger Cornea

Abnormality of
pupil

Glaucoma

Port wine stain
& Hypertrophy
of upper limb

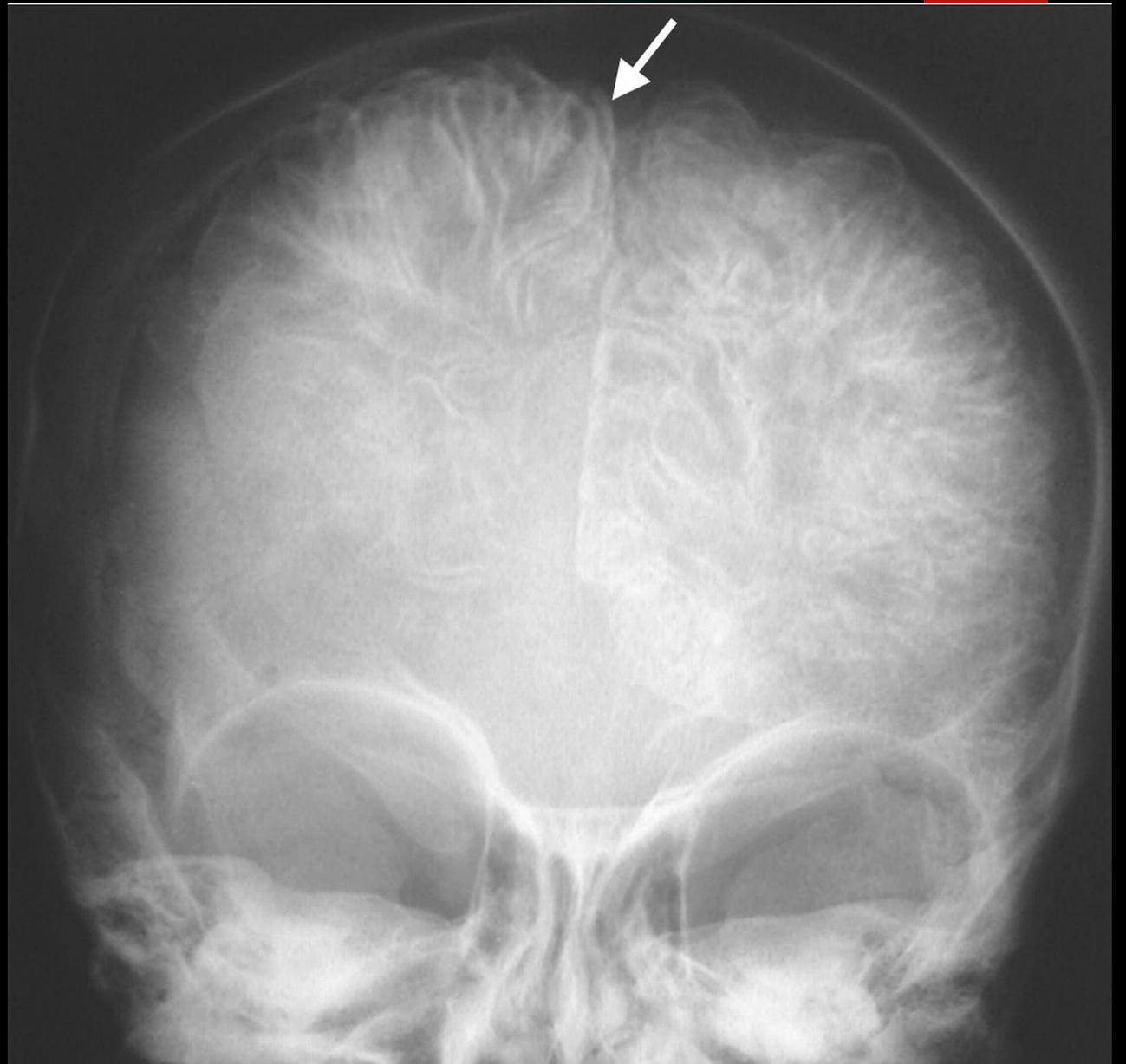


Combined
vascular
malformation of
capillaries veins
and lymphatics

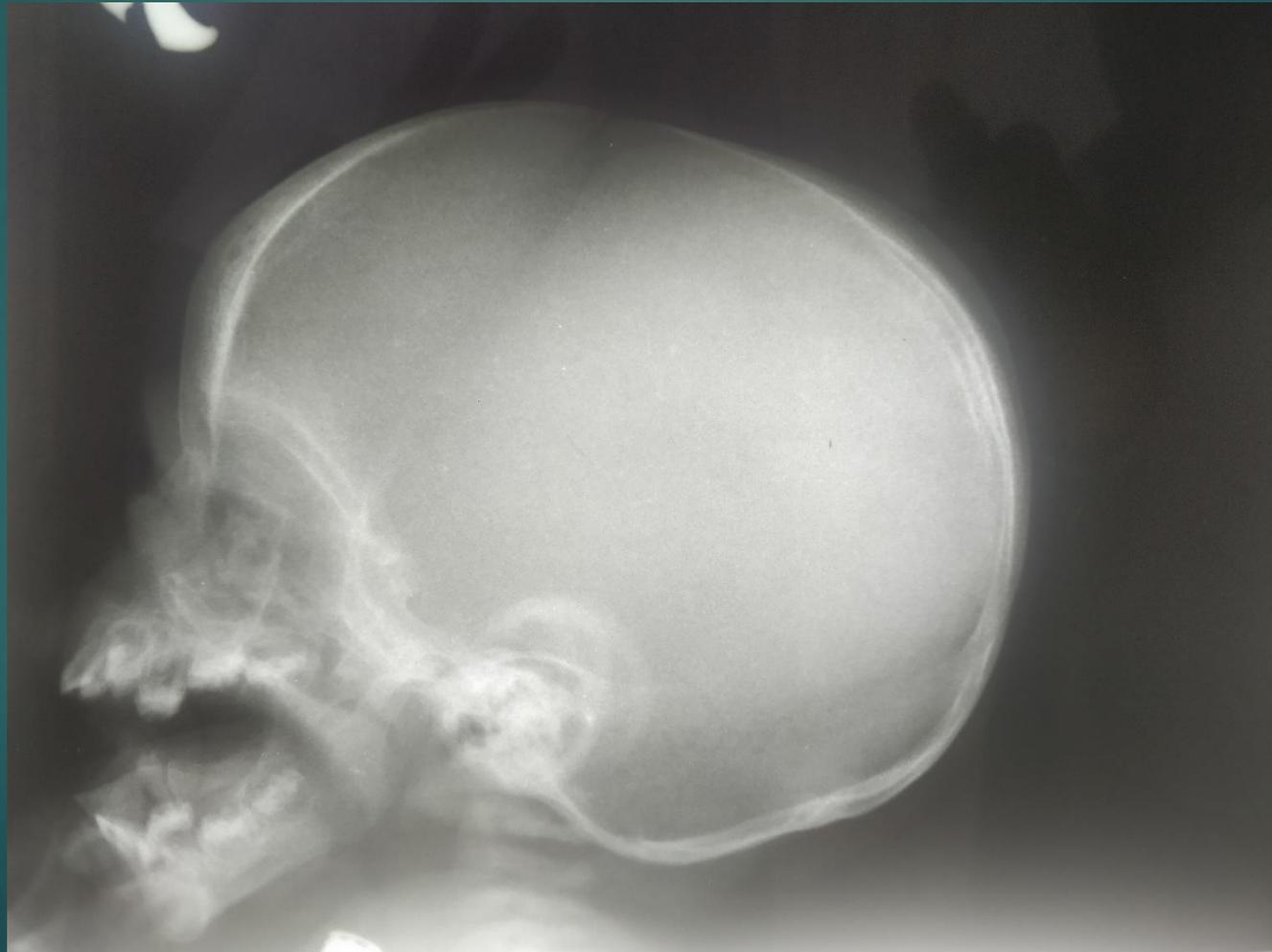
Limb
hypertrophy



Tram Track Sign

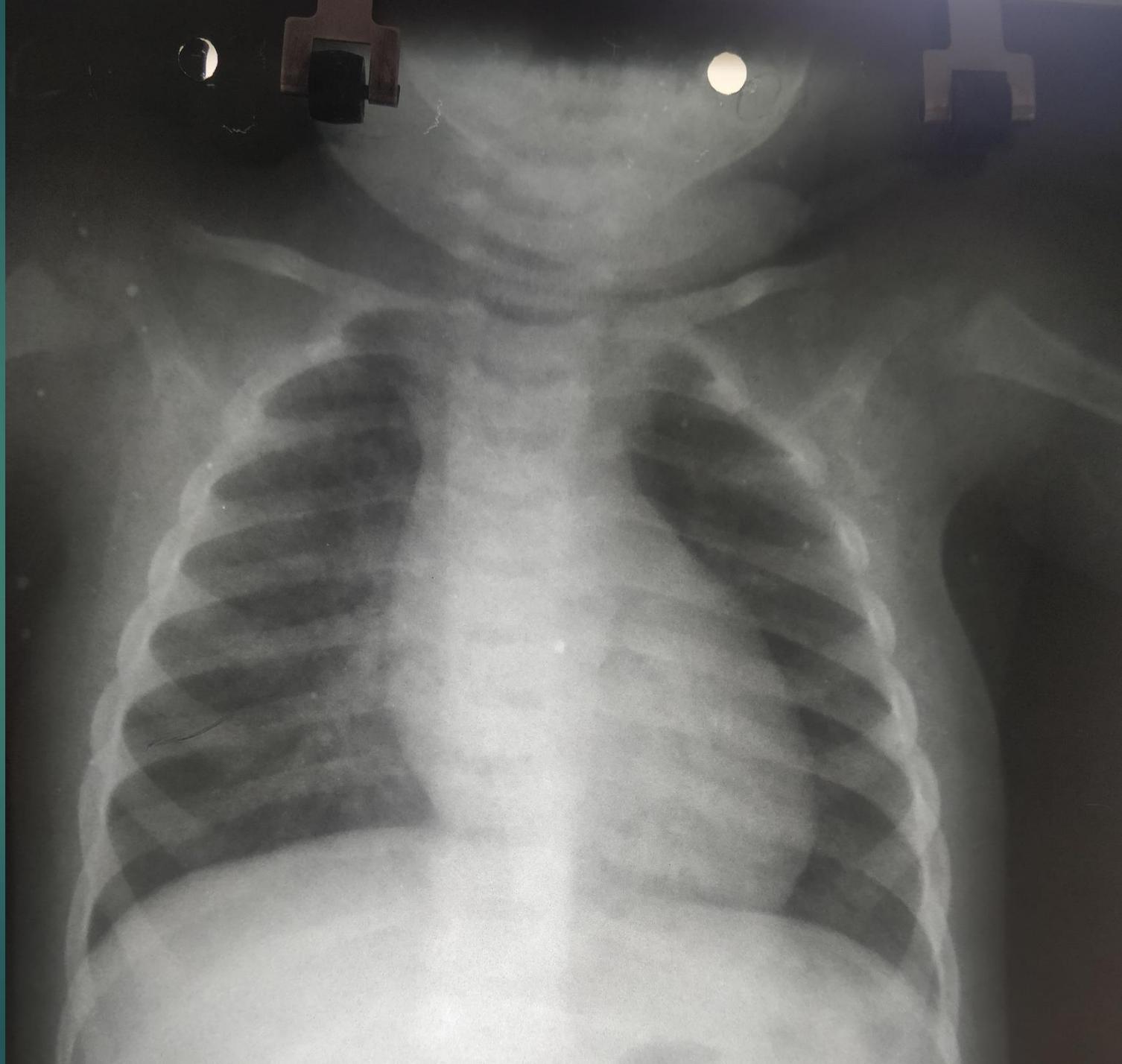


SKULL X-RAY



CHEST X-RAY

Cardiomegaly



Statistics

- ▶ Frequency in 1 to 20000 to 1 in 50000 birth
- ▶ **Mutation** in the GNAQ gene
- ▶ **SWS Type 1** -Facial and Leptomeningeal Angiomas
- ▶ **SWS Type 2** -Facial alone. No CNS
- ▶ **SWS Type 3** -Isolated Leptomeningeal angiomas

Presentation

- ▶ Also known as **Encephalotrigeminal Angiomatosis**
- ▶ Developmental Delay
- ▶ Cognitive impairment
- ▶ Seizures
- ▶ Paralysis
- ▶ 50% develop Glaucoma in Infancy

Sturge Weber Syndrome

Cardinal Features

Localized Cortical atrophy and calcifications.

“Tram Tracking”-Curvilinear double parallel lines on Plain Xray skull

Ipsilateral Port-Wine Facial Nevus,
1st division on Trigeminal.

Other Findings

- ▶ Ipsilateral exophthalmos/or **Glaucoma**
- ▶ Occulomeningeal **Capillary Hemangioma**
- ▶ Retinal Angiomas
- ▶ Convulsive **Seizures**

Treatment

- Anticonvulsants
- Lobectomy or Hemispherectomy for Refractory seizures
- Ophthalmology
- Physical Therapy
- Educational
- Genetic Profiling

Hemispherectomy

Anatomical
Functional

Klippel Trenaunay Syndrome

- ▶ Combined vascular malformation of capillaries veins and lymphatics
- ▶ Enlargement of bone, soft tissue of the limbs and asymmetric limb hypertrophy
- ▶ **Overlapping of Sturge Weber Syndrome plus Trenaunay Syndrome**

A very rare syndrome