Sturge-Weber Syndrome

Case Description
infant with reddish skin discoloration on one side of face present since birth

Image Description

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external photograph shows flat red pigmentation of the left side of facial skin consistent with facial port-wine stain; the left cornea is larger than the right cornea

Data acquisition
- History
  - age of onset/course
    - old photographs
  - signs of glaucoma ★
    - tearing
    - photophobia
    - blepharospasm
    - differences in eye size
    - intermittent corneal clouding
  - mental retardation, seizure
- Physical Exam
  - VA
  - IOP
  - ocular motility & alignment
    - eye preference
      - r/o amblyopia
  - slit lamp
    - conjunctival/episcleral vascular tortuosity
    - corneal diameter ★
    - corneal edema
    - Haab striae
    - iris heterochromia
  - funduscopy
    - diffuse choroidal hemangioma
    - serous RD
    - optic nerve cupping
  - cycloplegic refraction ★
    - myopia caused by long-standing glaucoma
  - limb hypertrophy, varicosities
    - Klippel-Trénaunay-Weber syndrome
  - ocular melanocytosis
    - phakomatosis pigmentovascularis

Additional Testing
- brain MRI or CT
  - r/o leptomeningeal angiomatosis

Assessment
- Sturge-Weber syndrome with port-wine stain

Treatment
- Medical
  - glaucoma
    - medical
      - topical glaucoma medication
        - avoid brimonidine in infants
          - can cause CNS depression, apnea
      - oral acetazolamide
    - Surgical
      - if medical rx not adequate
        - goniotomy
        - trabeculotomy
  - anisometropia correction
  - patching for amblyopia
  - PDT or radiation for symptomatic choroidal hemangioma
  - lasers can be used to decrease skin pigmentation
- Referrals
  - refer to neurologist for leptomeningeal angiomatosis

Patient Education/ Counselling
- Complications
  - discuss SW syndrome and its associations
    - glaucoma
      - 30%–70%
      - treatment of glaucoma associated with SW syndrome is difficult
        - often requires surgery
    - choroidal hemangioma
      - ≤50%
      - serous RD
    - leptomeningeal angiomatosis
- Follow-up
  - q6 mo
    - monitor for glaucoma
      - may occur at any time

Differential Diagnosis
- ***port-wine stain (nevus flammeus)*** associated with
  - ***Sturge-Weber syndrome (cerebrofacial angiomatosis)***
    - non-hereditary
    - nevus flammeus (portwine stain)
      - present from birth
      - usually unilateral
      - follows the distribution of CN V
    - glaucoma
      - particularly associated with upper lid hemangioma
    - parieto-occipital leptomeningeal hemangioma
      - seizure
      - mental retardation
    - heterochromia iridis
    - diffuse choroidal hemangioma
      - "tomato ketchup" appearance
      - exudative retinal detachment
  - ***Klippel-Trénaunay-Weber syndrome***
    - variant of cerebrofacial angiomatosis
      - non-hereditary
    - nonocular findings
      - nevus flammeus (portwine stain) of limb
      - hemihypertrophy of limb
        - bone & soft tissues
      - varicosities
      - intracranial hemangioma
    - ocular findings
      - nevus flammeus (portwine stain)
      - conjunctival telangiectasia
      - congenital glaucoma
  - ***phakomatosis pigmentovascularis***
    - in combination with ocular melanocytosis
- ***periocular contact dermatitis***
- ***preseptal cellulitis***
- ***erysipelas***

Ophthalmology Mind Maps
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