Aicardi Syndrome

Main Features

- Classic Triad:
  1. Infantile Spasms
  2. Agenesis of the corpus callosum
  3. Chorioretinal lacunae

Eye Findings

- Anterior Segment: Microcornea with or without coloboma
- Posterior Segment: Chorioretinal Lacunae radiating from optic disc most often
- absence of neurosensory retina, attenuated choroid, RPE hyperplasia
- Photoreceptor rosettes and photoreceptor inversions
- Visual prognosis better without lacunae
- Gross motor and language worse if many large lacunae

Etiology

- Xp22 mutation
- X-Linked Dominant
- Lethal in Males

Other Findings

- Neuro: Mental retardation, Characteristic EEG
- ENT: Cleft Lip and Palate
- Skin: Scalp lipomas, cavernous hemangiomas
- Bone: Vertebral and rib abnormalities

Reference

- OMIM 304050
- McMahon RG, Bell RA, Moore GRW, Ludwin SK. Aicardi's Syndrome, A Clinicopathologic Study. Arch Ophthalmol 102; 250-53,1984

syndrome