History
Female infant with seizure disorder.

Diagnosis
Aicardi Syndrome

Discussion
Aicardi syndrome is a rare neurodevelopmental disorder originally characterized by infantile spasms, callosal dysgenesis and chorioretinal lacunae. However, the phenotype of those affected can include many additional neurological and congenital expressions. Aicardi syndrome affects females, and, very rarely, males with a 47,XXY karyotype. Thus, Aicardi syndrome is thought to be caused by mutations in a gene on the X chromosome that causes death in XY males.
The study involved twenty-two females and one 47,XXY male all previously-diagnosed with Aicardi syndrome and presenting with the classic triad of infantile spasms, callosal dysgenesis and chorioretinal lacunae. Imaging studies noted polymicrogyria in all 23 (100%) subjects; 21 (91%) subjects showed greater anterior involvement, primarily in the frontal regions. Periventricular heterotopias were also present in 100% of the imaging studies, primarily around the body of the lateral ventricle.
Intracranial cysts can be seen in 20/21 (95%) imaging studies, of which 11 showed multiple cysts and 9 showed single cysts. 81% of cysts were midline interhemispheric, 29% were intraventricular, 10% were parenchymal and 8% extra-axial.
Of the 21 subjects whose posterior fossa could be examined, cerebellar abnormalities were present in 20. Abnormalities included superior foliar prominence of the vermis, interior vermin hypoplasia and dysplastic or hypo plastic cerebellar hemispheres.
The study did not assess callosal agenesis/dysgenesis in detail, but it is worth noting that agenesis was found in two-thirds of the imaging studies and dysgenesis in one-third.

Findings
MR-Sagittal T1 and axial T2 images show choroid plexus cysts, right perisylvian migrational disorder and marked colossal dysgenesis.

Reference

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