

Neurosciences Quiz

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A girl with recurrent tonic spasm

Clinical Presentation

A 7-month-old girl presented with a 4-week history of recurrent tonic spasms that occurred daily, predominantly upon awakening. She is able to roll over, however, is not sitting yet, and is not very attentive. Family history was unremarkable. Her head circumference was 40 cm, and she had no cutaneous signs. Neurologically, she had mild global hypotonia. No focal signs were noted.

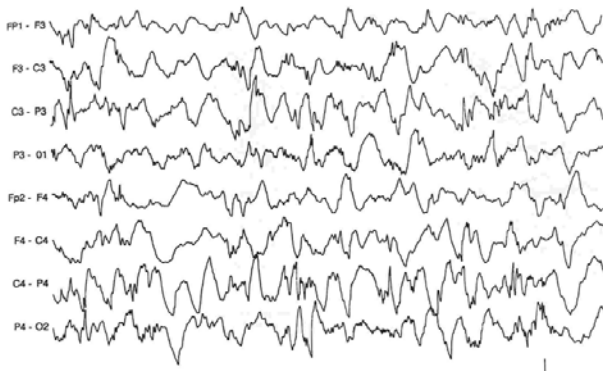


Figure 1 - Representative EEG sample.

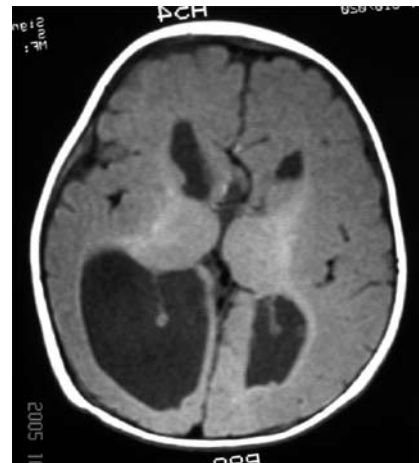


Figure 2 - Axial brain MRI.

Questions:

1. What are her EEG findings shown in Figure 1?
2. What are the abnormalities noted on her brain MRI shown in Figure 2?
3. What is the most likely underlying diagnosis?
4. Describe another important sign of this disorder?

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Answers

1. **EEG findings:** Chaotic pattern with high voltage slow delta waves intermixed with multifocal spikes, characterizing hypsarrhythmia.
2. **MRI abnormalities:** Agenesis of the corpus callosum, dilated posterior horns of lateral ventricles, and paucity of white matter posteriorly (colpocephaly).
3. **Diagnosis:** Aicardi syndrome presenting with infantile spasms.
4. **Another important sign of this disorder:** Retinal lacunae on fundal examination.

Discussion

Aicardi syndrome is a rare X-linked dominant disorder characterized by a triad of agenesis of the corpus callosum, infantile spasms, and chorioretinal lacunae.¹ The estimated prevalence is 2-15 per 100,000 girls.² Recently, Aicardi syndrome was linked to biallelic mutations in TREX1 at the AGS1 locus.³ TREX1 constitutes the major 3'→5' DNA exonuclease activity measured in mammalian cells. Most infants have severe psychomotor retardation and early onset intractable epilepsy. Infantile spasms were the most commonly observed, however, a variety of other seizure types were also reported.⁴ The prognosis is generally poor for seizure control and developmental progress.

References

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4. Rosser TL, Acosta MT, Packer RJ. Aicardi syndrome: spectrum of disease and long-term prognosis in 77 females. *Pediatr Neurol* 2002; 27: 343-346. Review.