

Cutaneous Lesions as a Clue to the Etiology of Extensive Intracranial Calcifications

Aicardi-Goutières Syndrome

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Figure Brain CT and Cutaneous Findings in Aicardi-Goutières Syndrome



(A–C) Brain CT reveals severe and extensive bilateral intracranial calcifications in the basal ganglia, thalamus, cerebellum, and subcortical white matter. (D) Dermatology examination uncovers small hypopigmented macules in the dorsal aspect of both hands.

A 29-year-old man presented with an 18-year history of progressive spastic gait and bizarre behaviors (Video 1). Cerebral palsy was initially suspected. Brain CT (Figure, A–C) showed extensive intracranial calcifications. Serum parathyroid hormone, calcium, and phosphate levels were normal. Hypopigmented macules were uncovered on the dorsal hands, which developed

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since age 6 (Figure, D). Genetic testing revealed a pathogenic heterozygous p.Gly1007Arg variant in *ADAR1*, confirming Aicardi-Goutières syndrome 6 (AGS6).¹

AGS is an autoinflammatory disease characterized by childhood-onset systemic inflammation with encephalopathy and cutaneous lesions due to enhanced type I interferon signaling.² Intracranial calcifications with cutaneous lesions should raise suspicion of AGS.

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Disclosure

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Appendix Authors

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Appendix (continued)

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