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

Aicardi-Goutières Syndrome

Yi-Heng Zeng, MD, Miao Zhao, MD, PhD, Xin-Xin Guo, MD, PhD, Ning Wang, MD, PhD, and Wan-Jin Chen, MD, PhD

Correspondence
Dr. Chen
wanjinchen75@fjmu.edu.cn

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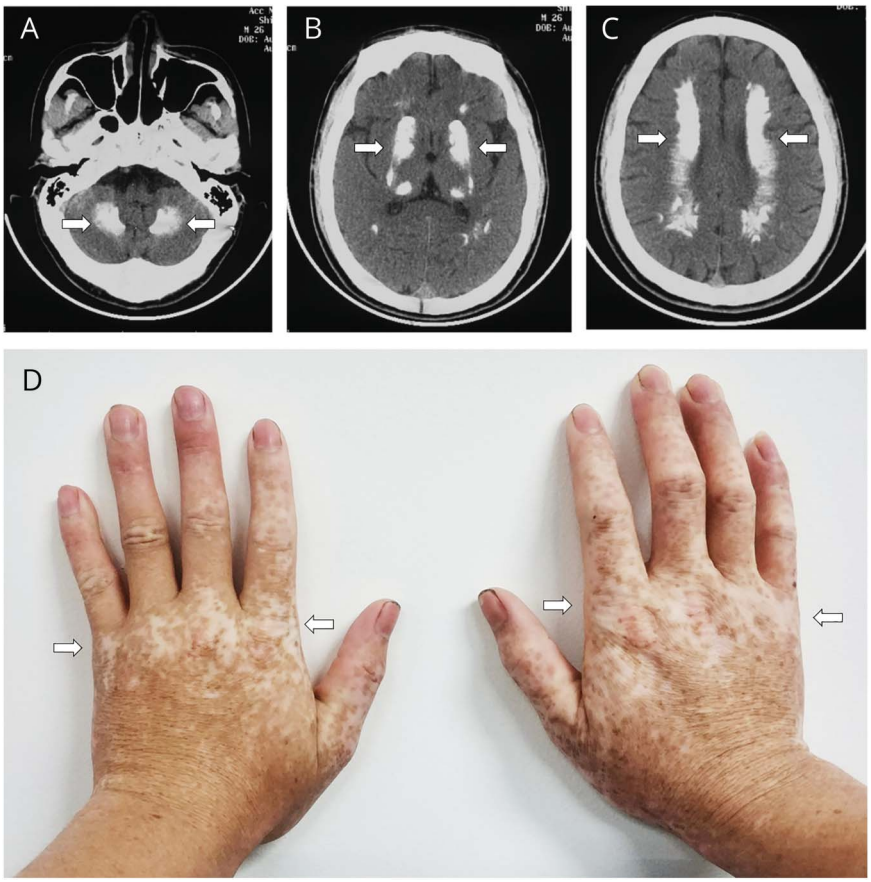
Figure Brain CT and Cutaneous Findings in Aicardi-Goutières Syndrome



(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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From the Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China.

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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

Name	Location	Contribution
Yi-Heng Zeng, MD	Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China	Drafting/revision of the manuscript for content, including medical writing for content; analysis or interpretation of data
Miao Zhao, MD, PhD	Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China	Major role in the acquisition of data
Xin-Xin Guo, MD, PhD	Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China	Major role in the acquisition of data; analysis or interpretation of data
Ning Wang, MD, PhD	Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China	Study concept or design
Wan-Jin Chen, MD, PhD	Department of Neurology and Institute of Neurology of First Affiliated Hospital, Institute of Neuroscience, and Fujian Key Laboratory of Molecular Neurology, Fujian Medical University, Fuzhou, China	Study concept or design

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