Cutaneous Lesions as a Clue to the Etiology of Extensive Intracranial Calcifications: Aicardi-Goutières Syndrome

Author(s):
Yi-Heng Zeng, MD\textsuperscript{1}; Miao Zhao, MD, PhD\textsuperscript{1}; Xin-Xin Guo, MD, PhD\textsuperscript{1}; Ning Wang, MD, PhD\textsuperscript{1}; Wan-Jin Chen, MD, PhD\textsuperscript{1}

This is an open access article distributed under the terms of the Creative Commons Attribution-NonCommercial-NoDerivatives License 4.0 (CC BY-NC-ND), which permits downloading and sharing the work provided it is properly cited. The work cannot be changed in any way or used commercially without permission from the journal.

Neurology® Published Ahead of Print articles have been peer reviewed and accepted for publication. This manuscript will be published in its final form after copyediting, page composition, and review of proofs. Errors that could affect the content may be corrected during these processes.
A 29-year-old man presented with 18 years’ 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 PTH, calcium, and phosphate levels were normal. Hypopigmented macules were uncovered on the dorsal hands which developed since the age of six (Figure, D). Genetic testing revealed a pathogenic heterozygous p.Gly1007Arg variant in ADARI, 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.

**Figure**: Brain CT and cutaneous findings in Aicardi-Goutières syndrome.

**Figure legend:**
(A, B and 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.
Video 1: The gait assessment of the 29-year-old AGS6 patient.
Video legend: The patient presented with spastic gait and forced laughter. Note the unremarkable cutaneous pigmented lesions on the dorsal aspects of hands.

Video 1-http://links.lww.com/WNL/B745

References
Cutaneous Lesions as a Clue to the Etiology of Extensive Intracranial Calcifications: Aicardi-Goutières Syndrome

Yi-Heng Zeng, Miao Zhao, Xin-Xin Guo, et al.

Neurology published online January 7, 2022
DOI 10.1212/WNL.0000000000013294

This information is current as of January 7, 2022