









Intracranial hemorrhage in a patient with Urbach-Wiethe disease

Hemorragia intracraniana em paciente com doença de Urbach-Wiethe

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A 39-year-old man presented with headache, left hemiparesis, and apathy. He also had epilepsy and hoarseness since infancy, tongue frenulum thickening, and yellow papular lesions

around his eyelids and fingers (→**Figure 1**). The computed tomography (CT) scan showed right frontal lobe hemorrhage, with “comma” shaped calcifications in both amygdalae,



Figure 1 Signs related to Urbach-Wiethe disease: (A) Dry skin and hyperkeratotic plaques on the left hand. (B) Thickened tongue frenulum. (C) Early alopecia on the top of the scalp and decreased hair density. (D) Beaded papular lesions on the eyelids, resembling a pearl necklace, also known as moniliform blepharosis.

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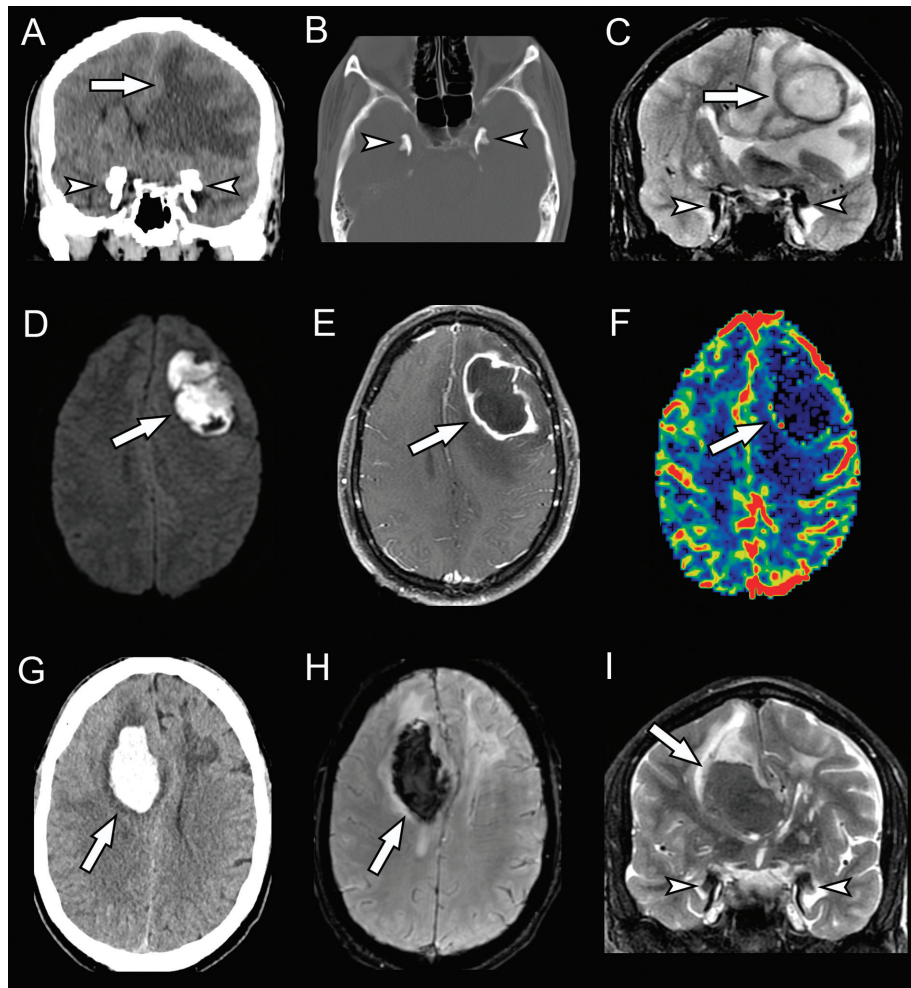


Figure 2 A noncontrast CT scan (A) demonstrates amygdalae comma-shaped calcifications (arrowheads) and hypoattenuation in left frontal lobe (arrow). A CT scan of the bone window (B) also shows calcifications (arrowheads). The coronal T2WI (C) depicts hypointense calcifications (arrowheads) and a lesion in left frontal lobe (arrow). Both DWI (D) and postcontrast T1WI (E) show central restricted diffusion and peripheral enhancement; no significant change in CBV color maps (F) is noted (arrows), suggesting an abscess. After 4 years the patient returned with an intraparenchymal hemorrhage in the right frontal lobe, seen in the noncontrast CT scan (G), SWI (H), and coronal T2WI (I) (arrows). Again, there are signs of calcification (arrowheads in I).

a characteristic finding of the Urbach-Wiethe disease (→ **Figure 2**). Previously, he presented with an intracranial pyogenic abscess due to poor dentition, perhaps related to hyaline deposits found along the parotid duct in these patients.¹ The whole exome sequencing shows a biallelic pathogenic variant c.816_817del (p.Cys272*) in the *ECM1* (→ **Figure 2**). Urbach-Wiethe disease is an autosomal recessive disorder, with hyaline-like material deposition in the skin, mucosae, and viscera.^{2,3} Neurological features include seizures, neuropsychiatric manifestations, intracranial hemorrhages, and calcifications in the mesial temporal lobes.⁴

Authors' Contributions

ALCCH: conceptualization, visualization, writing – original draft, writing – review & editing. GSAT: conceptualization, visualization, writing – review & editing. TTS: conceptualization, writing – original draft. LFF, JPMT: validation, writing – review & editing. GT: supervision, validation, writing – review & editing. FF: conceptualization, visualization,

supervision, writing – review & editing, and LTT: conceptualization, supervision, writing – review & editing.

Conflict of Interest

The authors have no conflict of interest to declare.

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