RESIDENT & FELLOW SECTION

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Mystery Case: Heidenhain variant of Creutzfeldt-Jakob disease

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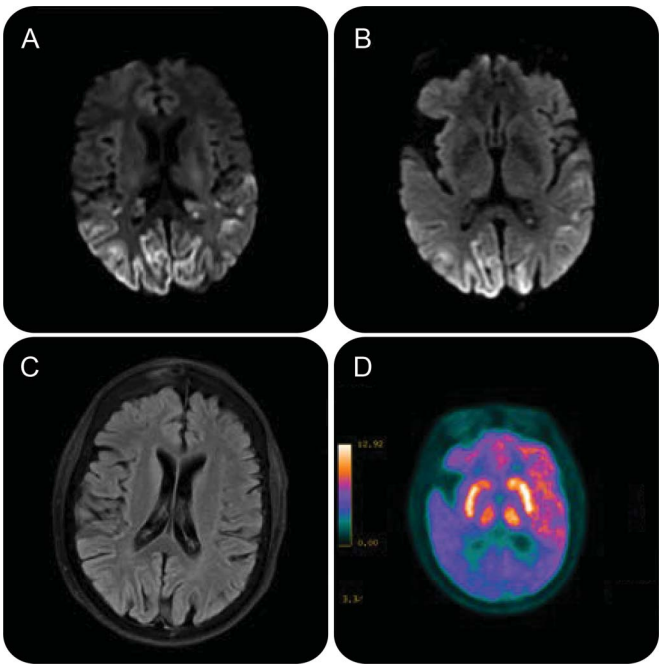
A 75-year-old woman complained of a "scrambled brain" for 1 month. She endorsed poor depth perception and an inability to construct "mental maps" of her home and the grocery store. Examination revealed impaired delayed recall, ocular apraxia, optic ataxia, and simultanagnosia (Bálint syndrome). Diffusion-weighted MRI demonstrated cortical hyperintensities in the occipital lobes extending into the right parietal lobe, suggesting spongiform encephalopathy (figure). The 14-3-3 protein and elevated neuron-specific enolase were detected in the CSF. The patient was diagnosed with the Heidenhain variant of Creutzfeldt-Jakob disease.[1] Early in the disease, this subgroup of patients with prion disease have isolated visual, not cognitive, symptoms and may be referred to an ophthalmologist.[2]

Figure Axial MRI and FDG-PET



Diffusion-weighted imaging shows prominent cortical hyperintensities in the occipital lobes (A) but not the basal ganglia (B). Fluid-attenuated inversion recovery signal intensity is mildly increased in the regions of restricted diffusion (C). FDG-PET shows diffuse hypometabolism involving the right parietal and bilateral occipital lobes (D).

AUTHOR CONTRIBUTIONS

Dr. Kalp: analyzed and interpreted data, drafted manuscript. Dr. Gottschalk: analyzed and interpreted data, revised manuscript.

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DISCLOSURE

The authors report no disclosures relevant to the manuscript. Go to Neurology.org for full disclosures.

REFERENCES

1. Kropp S, Schulz-Schaeffer WJ, Finkenstaedt M, et al. The Heidenhain variant of Creutzfeldt-Jakob disease. Arch Neurol 1999;56:55–61.
2. Cooper SA, Murray KL, Heath CA, Will RG, Knight RS. Isolated visual symptoms at onset in sporadic Creutzfeldt-Jakob disease: the clinical phenotype of the Heidenhain variant. Br J Ophthalmol 2005;89:1341–1342.

MYSTERY CASE RESPONSES

The Mystery Case series was initiated by the *Neurology*® Resident & Fellow Section to develop the clinical reasoning skills of trainees. Residency programs, medical student preceptors, and individuals were invited to use this Mystery Case as an educational tool. Responses were solicited through a group e-mail sent to the American Academy of Neurology Consortium of Neurology Residents and Fellows and through social media.

All of the responses we received came from individuals rather than groups. A total of 74% of respondents correctly identified the patient's constellation of symptoms as Bálint syndrome, and 42% noted that the classic localization for this syndrome is in the bilateral parieto-occipital regions. A total of 47% of respondents accurately identified Creutzfeldt-Jakob disease as the most likely diagnosis in light of this patient's clinical and radiographic presentation.

This case highlights a rare but well-described variant of Creutzfeldt-Jakob disease, and underscores the value of MRI in reaching this diagnosis.

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