RESIDENT & FELLOW SECTION

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Mystery Case: Cowden syndrome presenting with partial epilepsy related to focal cortical dysplasia

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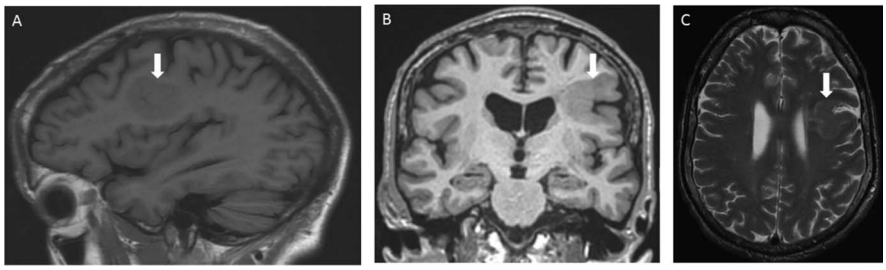
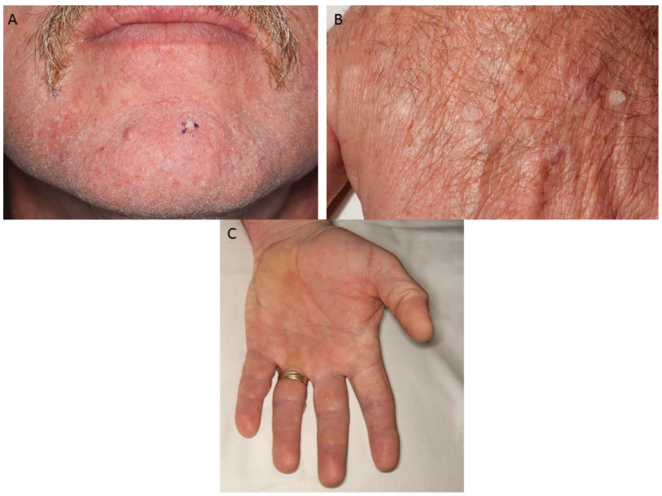
Figure 1 MRI brain



Sagittal T1-weighted MRI (A), oblique-coronal T1-weighted MRI (B), and axial T2-weighted MRI (C) show focal cortical dysplasia with abnormal sulcation involving the middle and inferior frontal gyrus posteriorly on the left with T2 signal extending to the ventricular surface thought to represent neuronal migration lines.

Figure 2 Skin changes found in Cowden syndrome



Skin examination revealed cobblestoning over the lower jaw (A), acral keratoses over the dorsal hands (B), and palmar pitting (C).

A 55-year-old man presented with seizures characterized by "tightening" of the right side and variable loss of awareness. EEG showed focal epileptogenic abnormalities over left and midline central regions. MRI showed left frontal focal cortical dysplasia (figure 1). He had multiple skin lesions (figure 2) and colonoscopy revealed gastrointestinal mucosal ganglioneuromas. Genetic testing of *PTEN* gene confirmed a diagnosis of Cowden syndrome (CS).

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CS is an autosomal dominant condition of hamartomas and tumors.[1] MRI is abnormal in 35%, commonly showing dysplastic gangliocytomas of cerebellum, meningiomas, and vascular malformations.[2] CS has not been reported presenting with partial epilepsy and focal cortical dysplasia.

AUTHOR CONTRIBUTIONS

N.D. Child and Dr. Cascino contributed to drafting and revising the manuscript. Dr. Cascino was responsible for the study concept.

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DISCLOSURE

N. Child reports no disclosures. G. Cascino serves as an Associate Editor for *Neurology*®. Go to Neurology.org for full disclosures.

REFERENCES

1. Farooq A, Walker LJ, Bowling J, Audisio RA. Cowden syndrome. Cancer Treat Rev 2010;36:577–583.
2. Lok C, Viseux V, Avril MF, et al. Brain magnetic resonance imaging in patients with Cowden syndrome. Medicine 2005;84:129–136.

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

From the Department of Neurology, Mayo Clinic, Rochester, MN.

of Neurology Consortium of Neurology Residents and Fellows and through social media. All the answers that we received came through social media, from individuals rather than groups. Most of the respondents (61%) indicated a putative diagnosis of tuberous sclerosis (TS) as the most likely etiology. Other considerations included focal cortical dysplasia, as well as demyelinating or infectious processes. The most complete answer came from Dr. Pedro Serrano Castro (Spain). In his response, he pointed out that the left parietal cortex is dysplastic and that the cutaneous lesions are suggestive of mTOR pathway dysfunction as seen in CS.

The cortical and subcortical tubers seen in TS have a different appearance on brain MRI with a low T1 and a high T2/fluid-attenuated inversion recovery signal. Other MRI diagnostic criteria for TS include the presence of subependymal nodules, subependymal giant cell astrocytomas, and cerebral white matter migration lines.

This Mystery Case illustrates the rare finding of a focal cortical dysplasia in a patient with CS.

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