

Adult diagnosis of Cockayne syndrome

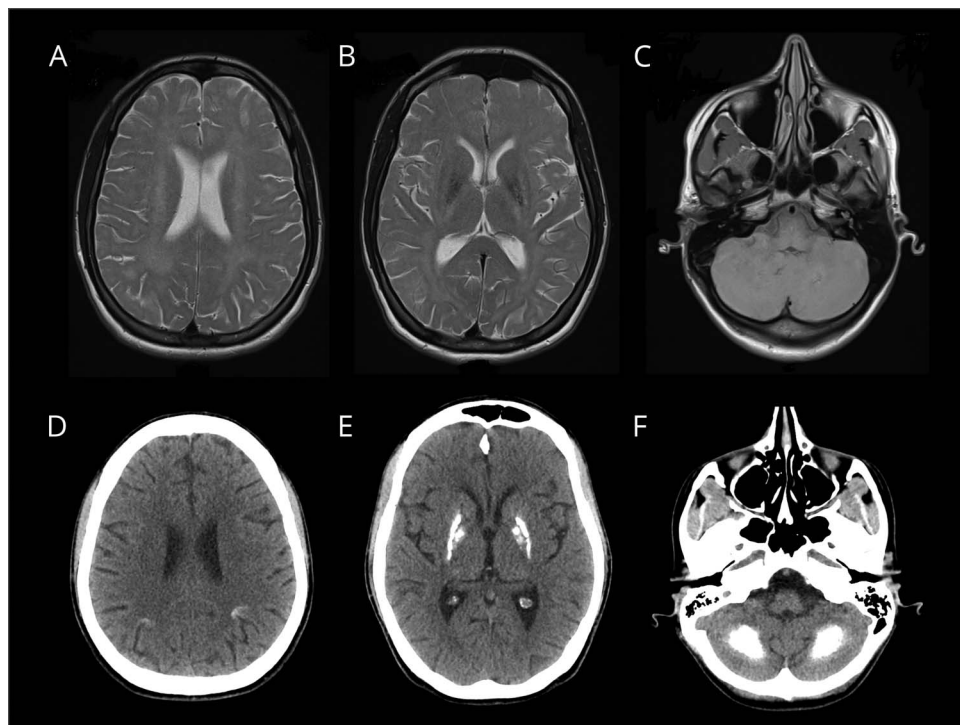
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Figure Axial T2-weighted MRI and axial CT scan



Axial T2-weighted MRI (A–C) shows white matter abnormalities and brain calcifications. Axial CT scan (D–F) demonstrates calcifications in the cortical sulci, basal ganglia, and cerebellum.

A 20-year-old woman presented with bilateral upper limb tremor, mild dystonia, and dysphonia (video). She had dermal photosensitivity, crowded facial features, short stature, and mild intellectual disability. Family history was unremarkable. Brain MRI and CT scan revealed subcortical white matter abnormalities and calcifications in the cortical sulci, basal ganglia, and cerebellum (figure). Genetic testing confirmed a Cockayne syndrome B with biallelic heterozygous mutations in the *ERCC6* gene (p.Gly715* in exon 10, p.Arg77* in exon 2). Cockayne syndrome should be suspected in adult patients when radiologic and clinical findings are consistent.^{1,2}

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Author contributions

A. Cocco: prepared the manuscript, images, and video. D. Calandrella: prepared the manuscript. M. Carecchio: critically revised the manuscript. B. Garavaglia: performed genetic analysis. A. Albanese: critically revised the manuscript and the video.

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Disclosure

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