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Teaching Neuro*Images*: The syndrome of cutaneous photosensitivity, growth failure, and basal ganglia calcification

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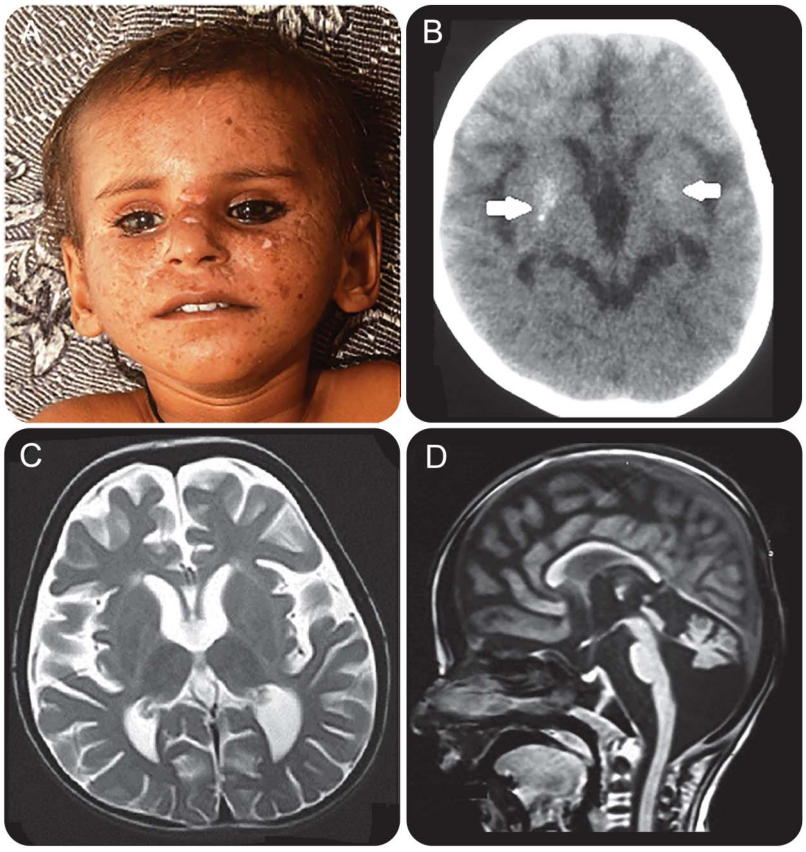
Figure Cockayne syndrome



(A) Facial appearance of the patient: progeroid appearance, sunken eyes, bird-like facies, photosensitive rash with areas of hyperpigmentation, and scarring. (B) Plain CT scan shows bilateral basal ganglia calcification (arrows) and diffuse brain atrophy. (C) MRI brain: T2-weighted axial section shows diffuse cerebral atrophy, hyperintensities in internal capsule, and periventricular white matter findings suggestive of hypomyelination with diffuse paucity of white matter most prominent in the parieto-occipital regions. (D) T1-weighted sagittal section shows diffuse cerebellar atrophy, enlarged cistern magna, atrophic appearance of pons and medulla, and thinned-out corpus callosum. Overall, the physical and radiologic findings are consistent with Cockayne syndrome.

A 2-year-old girl presented with growth failure, photosensitive rash, and developmental delay since infancy. On examination, she had a progeroid appearance, rash (figure, A), cachexia, microcephaly, and spasticity. Nerve conduction studies showed a demyelinating pattern and neuroimaging revealed basal ganglia calcification as well as cerebral and cerebellar atrophy (figure, B–D). Gene testing discovered homozygous *ERCC6* gene mutations (c.1874_1881delinsG in exon 9) in the child; her parents proved to be carriers.

Cockayne syndrome is an autosomal recessive neurodegenerative disorder characterized by progressive growth failure, microcephaly, intellectual impairment, retinal pigmentary degeneration, and photosensitivity due

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to ultraviolet-induced DNA repair defect.[1] Clinicians should identify the characteristic appearance and radiologic features; the causative genes are *ERCC6* and *ERCC8*.[2]

AUTHOR CONTRIBUTIONS

Arushi Gahlot Saini: original drafting and revising the manuscript for intellectual content. Naveen Sankhyan: critical review of the manuscript for intellectual content. Sameer Vyas: analysis and interpretation of radiologic data and critical review of the manuscript. Vincent Laugel and Nadege Calmels: genetic analysis and critical review of the manuscript. Pratibha Singhi: clinician-in-charge, conceptualization of the study, and critical review of the manuscript for intellectual content.

STUDY FUNDING

No targeted funding reported.

DISCLOSURE

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

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Neurology 2016;87;e56-e57
DOI 10.1212/WNL.0000000000002949