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Teaching Neuro*Images*: Griscelli syndrome and CNS lymphohistiocytosis

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A 3-year-old boy developed viral illness followed by fever, altered sensorium, focal seizures, and neuroregression. Examination showed silvery-gray hair (figure 1A), bilateral papilledema, spastic quadriparesis, brisk muscle-stretch reflexes, extensor plantars, hepatosplenomegaly, and normally pigmented skin, iris, and retina. Hair microscopy confirmed Griscelli syndrome (GS) (figure 1, B–D). MRI brain was suggestive (figure 2, A–D). CSF showed 20 degenerated leukocytes. He died of an intercurrent illness 2 months later.

GS is an autosomal recessive disorder of melanosome transport with hypopigmentation, immunodeficiency, and early death.[1] Silvery-gray hair is an important clinical clue.[2] The index case was probably GS type 2. Fatal complications include "accelerated phase" characterized by lymphohistiocytic proliferation in various organs, including brain.

AUTHOR CONTRIBUTIONS

Arushi Gahlot Saini: draft of manuscript and review of literature. R. Nagaraju: contribution to the draft of the manuscript. Jitendra K. Sahu: critical review of manuscript for important intellectual content and final approval of the version to be published. Amit Rawat: analysis of hair microscopy, critical review of manuscript, and final approval of the version to be published. Sameer Vyas: analysis of the radiologic data, critical review of manuscript, and final approval of the version to be published. Pratibha Singhi: Clinician-in-charge, concept and design of the study, critical review of manuscript for important intellectual content, and final approval of the version to be published.

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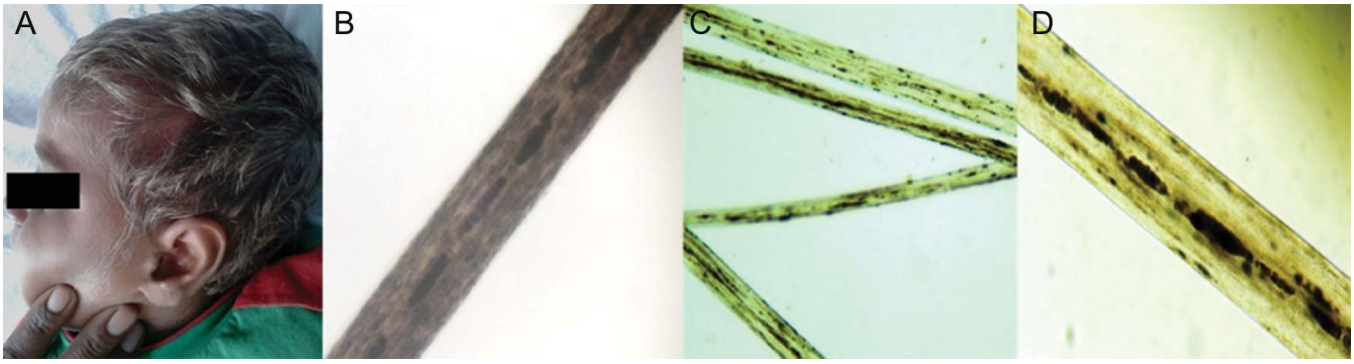
Figure 1 Facial and scalp hair characteristics of Griscelli syndrome



(A) Clinical photograph of the face showing silvery-gray hair over the scalp and eyebrows. (B) Photomicrographs of normal hair shafts on routine light microscopy. (C, D) Photomicrographs of hair shaft of index patient show hypopigmentation along with large and irregular melanin granules.

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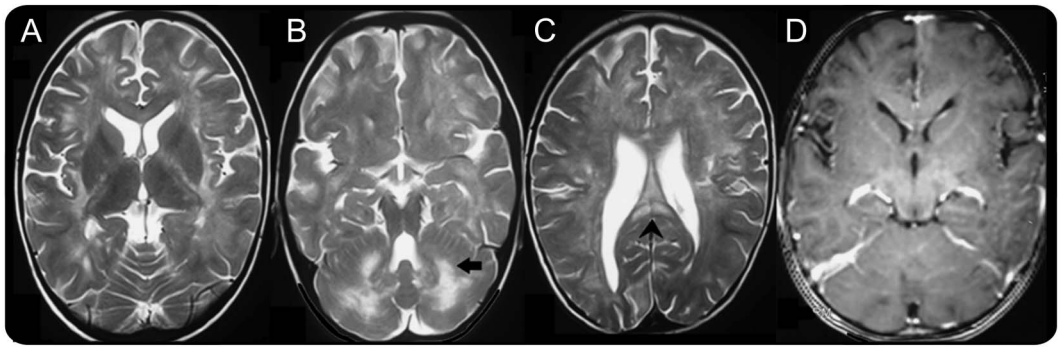
Figure 2 Neuroimaging



MRI of the brain: axial T2 images show bilateral, symmetrical, diffuse hyperintensities involving (A) subcortical white matter, bilateral external capsule, posterior limb of bilateral internal capsule with sparing of subcortical U-fibers, (B) bilateral cerebellar hemispheres (arrow), and (C) splenium of corpus callosum (arrowhead). (D) Axial T1 images with gadolinium contrast administration did not show any contrast enhancement. The overall picture is consistent with lymphohistiocytic infiltration of the brain.

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