

Teaching NeuroImages: Griscelli syndrome and CNS lymphohistiocytosis

Arushi Gahlot Saini, MD
S. Nagaraju, MBBS
Jitendra Kumar Sahu,
MD, DM
Amit Rawat, MD
Sameer Vyas, MD
Pratibha Singhi, MD

Correspondence to
Dr. Singhi:
doctorpratibhasinghi@gmail.com

Download teaching slides:
Neurology.org

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.

STUDY FUNDING

No targeted funding reported.

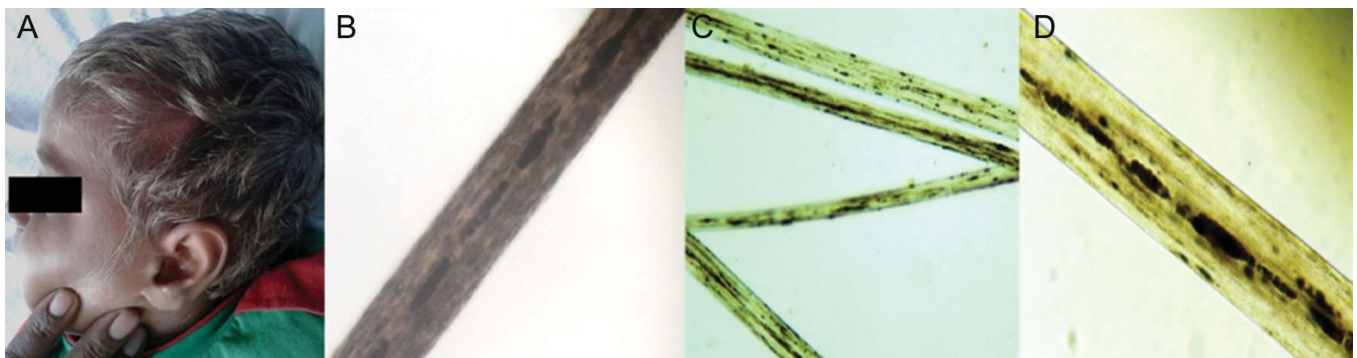
DISCLOSURE

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

REFERENCES

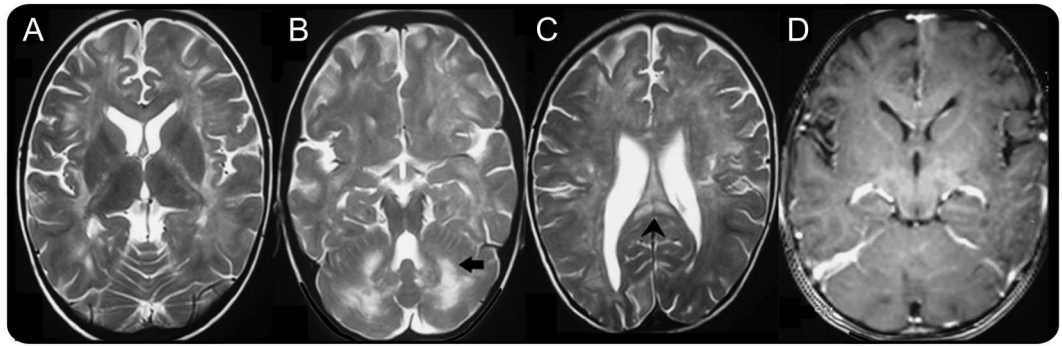
- Masri A, Bakri FG, Al-Hussaini M, et al. Griscelli syndrome type 2: a rare and lethal disorder. *J Child Neurol* 2008;23:964–967.
- Manglani M, Adhvaryu K, Seth B. Griscelli syndrome: a case report. *Indian Pediatr* 2004;41:734–737.

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.

From the Unit of Pediatric Neurology and Neurodevelopment (A.G.S., P.S.), Department of Pediatrics (S.N., J.K.S., A.R.), and Department of Radiodiagnosis (S.V.), Postgraduate Institute of Medical Education and Research, Chandigarh, India.



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.

Neurology®

Teaching *NeuroImages*: Griscelli syndrome and CNS lymphohistiocytosis

Arushi Gahlot Saini, S. Nagaraju, Jitendra Kumar Sahu, et al.

Neurology 2014;82:e122-e123

DOI 10.1212/WNL.0000000000000288

This information is current as of April 7, 2014

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/82/14/e122.full
Supplementary Material	Supplementary material can be found at: http://n.neurology.org/content/suppl/2014/04/05/82.14.e122.DC1
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/82/14/e122.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Clinical Neurology http://n.neurology.org/cgi/collection/all_clinical_neurology All Genetics http://n.neurology.org/cgi/collection/all_genetics All Imaging http://n.neurology.org/cgi/collection/all_imaging Leukodystrophies http://n.neurology.org/cgi/collection/leukodystrophies MRI http://n.neurology.org/cgi/collection/mri
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright © 2014 American Academy of Neurology. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

