Teaching NeuroImages: Nasu Hakola syndrome

Kshitij Mankad, MRCP,
FRCR
Indran Davagnanam,
FRCR

Address correspondence and
reprint requests to Dr. Kshitij
Mankad, The National Hospital
for Neurology and Neurosurgery,
Queen Square, London WC1N
3BG, UK
drmankad@gmail.com

Figure 1 Right distal femoral pathologic fracture on a background of cystic bones

A Sri Lankan man in his 40s presented with a femoral fracture (figure 1) resulting from minor trauma. History from family members suggested progressive cognitive decline. Examination revealed intermittent euphoria, memory impairment, and apraxia. He was doubly incontinent with preserved primitive reflexes. CT (figure 2) revealed atrophy with frontal predilection and calcified basal ganglia.

Figure 2 CT brain scan revealing atrophy, ventricular dilation, and dense basal ganglia calcification (arrows)

A diagnosis of Nasu Hakola syndrome (polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy) was considered.

This condition is characterized by a combination of frontal lobe dementia and widespread cystic destruction of bones.¹ It results from mutations in the DAP-12 or TREM-2 surface signaling genes.²

REFERENCES

1. Paloneva J, Raininko R, Partanen J, et al. CNS manifestations of Nasu-Hakola disease: a frontal dementia with bone cysts. *Neurology* 2001;56:1552–1558.
2. Klunemann HH, Ridha BH, Magy L, et al. The genetic causes of basal ganglia calcification, dementia, and bone cysts: DAP12 and TREM2. *Neurology* 2005;64:1502–1507.

From The National Hospital for Neurology and Neurosurgery, Queen Square, London, UK.

Disclosure: The authors report no disclosures.

Neurology®

Teaching *NeuroImages*: Nasu Hakola syndrome

Kshitij Mankad and Indran Davagnanam

Neurology 2010;74:e102

DOI 10.1212/WNL.0b013e3181e24295

This information is current as of June 7, 2010

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/74/23/e102.full
References	This article cites 2 articles, 2 of which you can access for free at: http://n.neurology.org/content/74/23/e102.full#ref-list-1
Citations	This article has been cited by 1 HighWire-hosted articles: http://n.neurology.org/content/74/23/e102.full##otherarticles
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): All Cerebrovascular disease/Stroke http://n.neurology.org/cgi/collection/all_cerebrovascular_disease_stroke All Clinical Neurology http://n.neurology.org/cgi/collection/all_clinical_neurology All Cognitive Disorders/Dementia http://n.neurology.org/cgi/collection/all_cognitive_disorders_dementia All Movement Disorders http://n.neurology.org/cgi/collection/all_movement_disorders
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 . All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.