

Teaching NeuroImages: Nasu Hakola syndrome

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



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



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.

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

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