Nasu-Hakola Disease Revealed on X-ray

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We encountered a 37-year-old woman who had experienced frontal lobe symptoms since her twenties without a history of pathological fractures or bone pain. Brain MRI images showed frontotemporal atrophy and leukoencephalopathy (Picture A). Her clinical history and imaging findings prompted us to take X-rays of the extremities. The X-ray images revealed multiple bone cysts in the distal extremities, thus indicating Nasu-Hakola disease (NHD) (Picture B). Genetic testing confirmed a mutation in the DAP12 gene (a single base mutation T to C in the start codon of exon 1) specific to NHD (1). X-ray is a very useful tool for diagnosing NHD, even in patients without obvious bone symptoms (2). Obtaining X-rays of the extremities may help to diagnose early-onset frontotemporal dementia.

The authors state that they have no Conflict of Interest (COI).

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