

Computed tomography and magnetic resonance imaging in the osseous phase of Nasu-Hakola disease

Tomografia computadorizada e ressonância magnética na fase óssea da doença de Nasu-Hakola

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Nasu-Hakola disease (NHD-polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy; PLOSL) is a rare autosomal recessive disorder, caused by mutations in two genes: *TREM 2* and *DAP 12*. NHD is characterized by a combination of diffuse bone cysts and pre-senile dementia. Most of the NHD cases first present in early adulthood with skeletal abnormalities (osseous phase). Neurological symptoms manifest in the fourth decade of life as prefrontal syndrome^{1,2,3}. A 32 year-old male patient presented bone fractures, usually after minor traumas and cystic lesions on X-Rays (Figure 1). He denied any personality change or memory disturbances. The neurological examination was normal. The neuropsychological tests displayed only easy distractibility. CT and MRI demonstrate abnormalities in the basal ganglia and white matter (Figure 2) showing that imaging findings precedes neuropsychiatric symptoms.



Figure 1. R-xays of the right hand demonstrates multiple cystic lesions in the carpal bones.

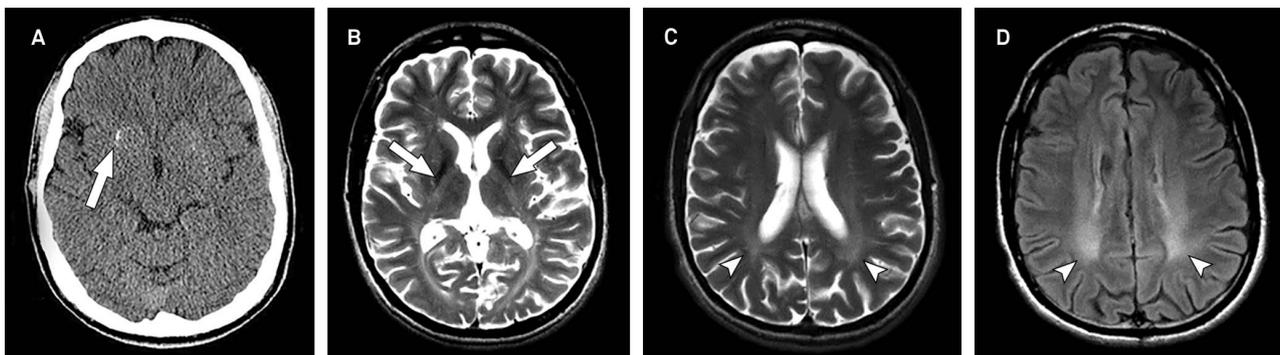


Figure 2. Head CT discloses punctate basal ganglia calcification (arrows in A). Brain MRI, axial T2-weighted (B, C) and axial FLAIR (D) images show diffuse cerebral atrophy, bilateral and symmetric hyperintensity in the posterior limbs of the internal capsules (arrows in B), and in the parieto-occipital periventricular white matter and centrum semiovale (arrowheads in C and D).

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