Pseudoxanthoma elasticum presenting as akinetic-rigid parkinsonism and dementia

A 54-year-old man presented with a three-year history of progressive cognitive decline, abulia, apathy, slowing movements and global rigidity. Medical history, confirmed by skin biopsy, revealed a definite diagnosis of pseudoxanthoma elasticum. Examination disclosed angioid streaks at funduscopic evaluation, skin lesions (Figure 1), bradykinesia and marked symmetrical akinetic-rigid parkinsonism with cogwheel rigidity. Neuroimaging studies showed multiple lacunar infarcts in the basal ganglia and brainstem. Tc-99m SPECT imaging showed bilateral frontal hypoperfusion (Figure 2).

Pseudoxanthoma elasticum is a rare autosomal recessive ectopic mineralization disorder, which can present a complex neurological picture due to severe vascular compromise, rarely with movement disorders.

Figure 1. Typical skin lesions in pseudoxanthoma elasticum. Note the presence of (A, B) axillary and neck yellowish papules and plaques with cobblestone appearance and (C) mucosal lesions in the inner aspect of the lower lip (white arrow).

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References


Figure 2. Neuroimaging findings in pseudoxanthoma elasticum. Axial brain MR imaging showing multiple pontine and bilateral basal ganglia lacunar infarcts with iron deposition in FLAIR sequence (A–C), T2-weighted imaging (D–F), T1-weighted imaging (G) and SWI sequences (H). (I,J) Brain Tc-99m SPECT disclosing bilateral frontal hypoperfusion (white arrows).