Neurological Case Reports

Clinical Image
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The Pulvinar Sign from Fabry Disease

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Clinical Image

A 33-year-old woman presented in the emergency department with sudden onset axial and left appendicular in coordination and dysarthria. During this episode, she was hospitalized and an extensive clinical and complementary research was carried out. Although laboratorial analysis were normal, magnetic resonance imaging of the brain showed pre contrast symmetric hyper intensity of both globus pallidus and thalamus, predominating in pulvinar nuclei (Figure). Her medical history included cochlear nerve dysfunction, angiokeratoma of the skin, acroparesthesia and abdominal pain. Family history includes paternal death after acute myocardial infarction at 32 years of age. Genetic sequence analysis for Fabry diseasegene showed a heterozygous mutation in exon 2 (p.R118C). Fabry Disease (FD; Online mendelian inheritance in man #301500) is a rare, X-linked inherited, progressive, multisystem lysosome storage disorder, resulting in α-galactosidase. A deficiency and progressive accumulation of globotriaosylceramide in various tissues, including, the pulvinar region of the thalamus. Clinical manifestations in this female patient are interpreted as components of X-inactivation [1,2]. Enzyme replacement therapy with humanized recombinant α-Gal A was instituted and the neurological condition was stabilized [3].

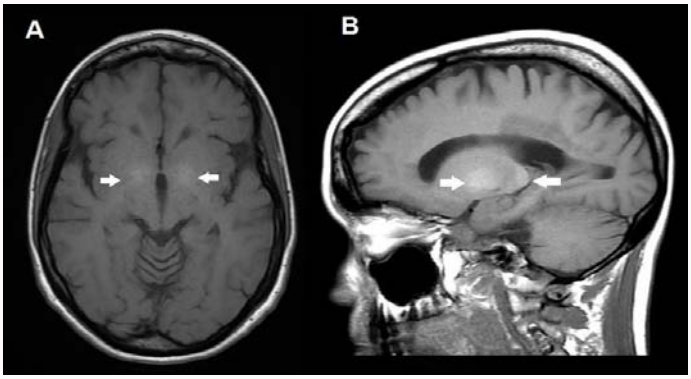

Figure: Magnetic resonance imaging of the brain showed precontrastT1 axial (Panel A, arrow) and sagittal (Panel B, arrows) symmetric hyper intensity of both globus pallidus and thalamus, predominating in pulvinar nuclei.

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