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IMAGES IN NEUROLOGY

Nodular heterotopia: a rare finding in patients with epilepsy and SCN1A mutation

Heterotopia nodular: um achado raro em pacientes com epilepsia por mutação no gene SCN1A

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A 25-year-old man had started febrile seizures from 6 months of age and developed important neurodevelopmental delay. At four years presented with status epilepticus and sequentially with refractory epilepsy. He presents with motor delay, behavior disorder and absent expressive language. Brain MRI showed periventricular nodular heterotopias (Figures 1 and 2). The Next Generation Sequencing (NGS) came with a rare frameshift mutation in heterozygosis for SCN1A gene.

The presence of periventricular heterotopias in association with SCN1A mutation is rare[1]. In literature, we found only two cases reported, being it the most common finding in patients with Filamin A gene (FLNA)[1,2]. The CNS anomalies more common in SCN1A mutation are cortical atrophy, cerebellar atrophy, white matter hyperintensity, ventricular enlargement, hippocampal sclerosis, or cortical dysplasia[3,4].

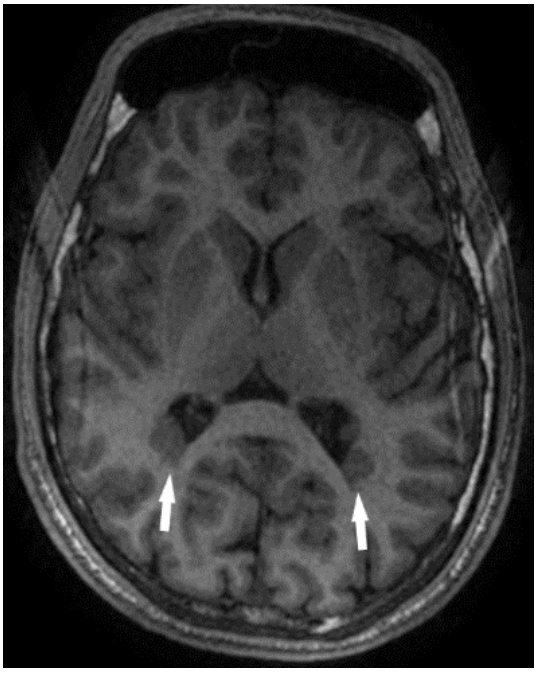

Figure 1. Axial T1WI with nodules of grey matter in the trigones of the lateral ventricles, and normal nuclei of deep gray matter.

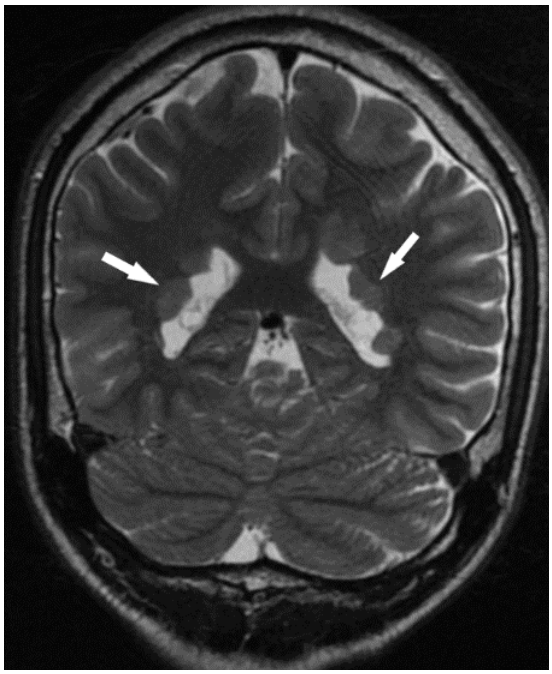

Figure 2. Coronal T2WI with nodules of grey matter in the trigones of the lateral ventricles.

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