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Widening the spectrum of LAMA 2 congenital muscular dystrophy (MDC1A): cobblestone malformation

Ampliação do espectro da distrofia muscular congênita associada ao LAMA 2 (MDC1A): malformação do tipo "cobblestone"

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A 4-year-old boy with *LAMA2*-related congenital muscular dystrophy had two pathogenic variants (NM_000426): c.1255delA and c.2461A>C. Magnetic resonance imaging (MRI) of the brain showed signal abnormalities in supratentorial white matter (WM), which are conspicuous findings in this disease[1]. Interestingly, MRI also depicted malformations of cortical development - symmetric bilateral parieto-occipital bumpy or pebbly cortical surface (cobblestone malformation)[2] (Figure 1).

This report expands *LAMA2*-related radiological phenotype to include not only WM abnormalities, but also predominantly posterior cerebral cortex changes.

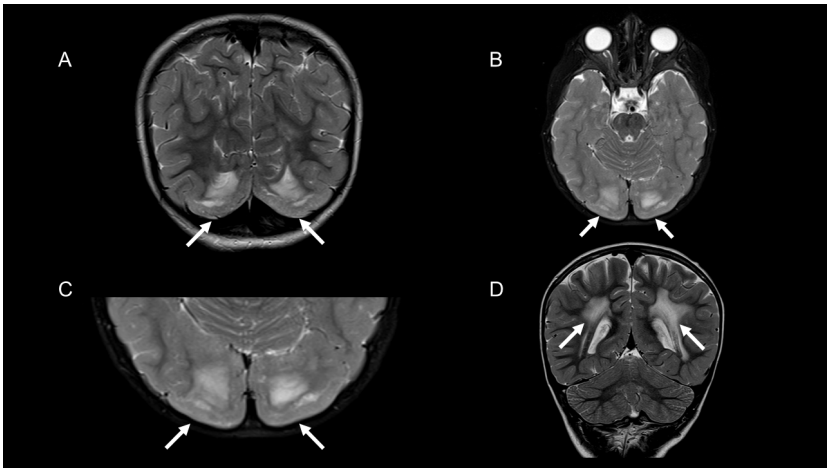

Figure 1. (A) Coronal T2-weighted image (T2WI) showing bilateral and symmetric type II lissencephaly or "cobblestone" lissencephaly (arrows). (B) Axial T2-weighted image (T2WI) showing bilateral and symmetric type II lissencephaly or "cobblestone" lissencephaly (arrows). (C) Axial T2-weighted image (T2WI) with a closer view of the symmetric type II lissencephaly or "cobblestone" lissencephaly (arrows), in contrast with normal cortical development. (D) Coronal T2-weighted image (T2WI) showing signal abnormalities in the periventricular white matter (arrows).

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