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Megalencephalic leukoencephalopathy with subcortical cysts (MLC) – a case with clinical and magnetic resonance imaging (MRI) dissociation

Leucoencefalopatia megalencefálica com cistos subcorticais (MLC) – um caso com dissociação clínica e de resonância magnética

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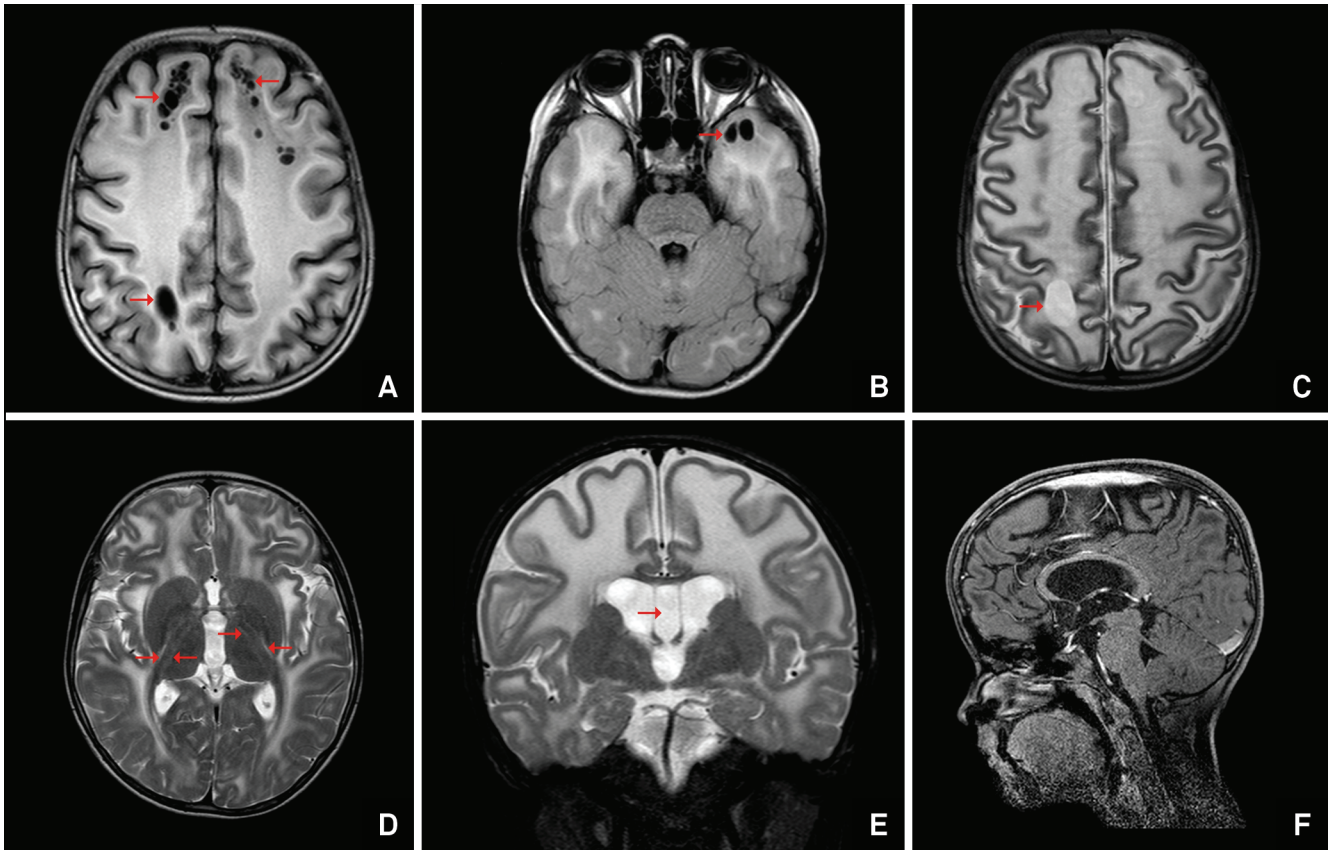

Figure 1. (A), Axial T1-weighted image shows diffuse cerebral white-matter abnormalities with frontoparietal subcortical cysts (arrows). (B), Axial FLAIR image exhibits subcortical cysts (arrows) in the anterior temporal lobe intermingled with diffuse white-matter hyperintensity. (C) and (D), Axial T2-weighted images with diffuse white-matter abnormalities, the external and extreme capsules are involved and the posterior limb of the internal capsule shows a double-line of abnormal intensity (arrows in D). (E), Coronal T2-weighted image highlights a diffuse brain swelling with white-matter hyperintensity and persistence of the cavum septum pellucidum (arrow). (F), Sagittal T1-weighted image with fat suppression and after endovenous gadolinium contrast, note the cranio-facial disproportion without abnormal enhancement. The corpus callosum is relatively spared.

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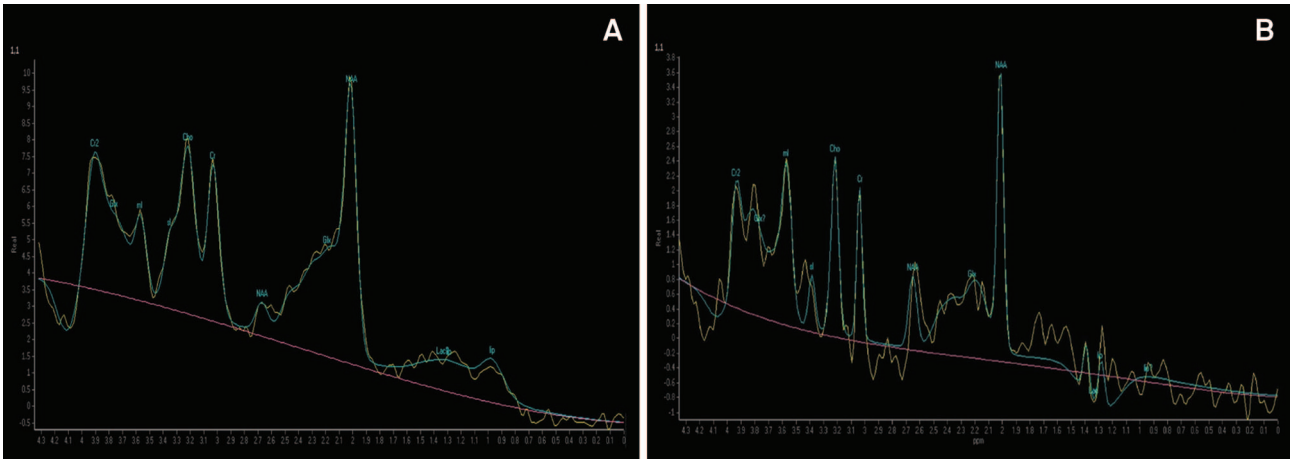

Figure 2. Short TE (31ms) proton-spectroscopy in a control (A) and in the patient with MLC (B) demonstrates mild increased concentration of choline (Cho, in 3.24 ppm) and myoinositol (mI in 3.56 ppm), there is also mild decreased NAA (this pattern is compatible with mild disease, indicating gliosis and rarefaction of the white-matter). In patients with another spectrum of this disease (severe neurological deterioration), the white-matter is totally cystic and proton-spectroscopy shows peaks of lactate (in 1.33 ppm) and glucose (in 3.4 and 3.8 ppm).

A ten-year-old boy with macrocephaly. His head circumference soon after birth started growing above normal. Developmental milestones were normal and neurological examination showed mild hypotonia in the first years of life. The only complain is frequent falls, without cerebellar or motor abnormalities on neurological examination. MLC diagnosis was based on MRI findings (Figures 1 and 2). The absence of cerebellar abnormalities in this case is useful to rule out congenital muscular dystrophy in patient with macrocephaly[1].

MLC is associated with MCL1 gene mutation in 70% of the cases[2], leading to disbalanced intracelullar ion concentration and astrogliosis[3,4].

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