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Acute hyperammonemic encephalopathy secondary to ornithine transcarbamylase deficiency in a child: findings in diagnostic imaging

Encefalopatia hiperamonêmica aguda secundária à deficiência de ornitina transcarbamilase em uma criança: achados em exames de diagnóstico por imagem

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A four-year-old girl presented with disorientation, drowsiness, anisocoria, tetraparesis and sialorrhea. She had shown significant behavioral changes over the previous six months. Computed tomography revealed bilateral involvement of the frontotemporoparietal and insular cortices, deep and subcortical white matter, confirmed by magnetic resonance imaging, which also showed signs of water diffusion restriction, hyposignal in T1-weighted sequence and no gadolinium enhancement (Figure). A prominent lactate peak was observed in spectroscopy. Urea cycle disorder was suspected after serum urea level was found to be 11 times higher than normal, which was confirmed after a genetic test was positive for heterozygous ornithine transcarbamylase deficiency[1,2,3,4,5].

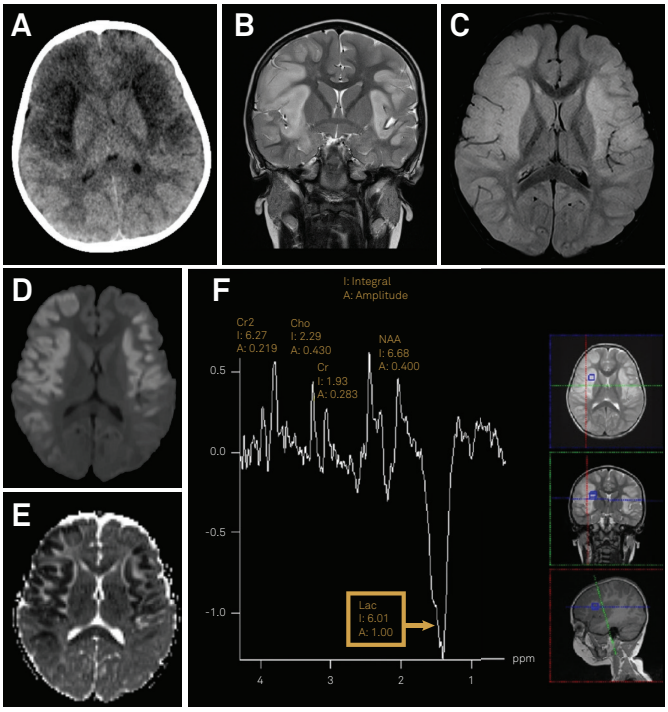

Figure. Computed tomography (A), showing extensive hypodensity affecting the deep and subcortical cerebral white matter mainly in areas of frontal, temporal, parietal and insular lobes bilaterally, predominantly symmetrical. Brain magnetic resonance imaging (B,C,D,E,F), showing in T2-weighted sequence (B) and FLAIR (C) a hyperintensity of the cortex, deep and subcortical white matter, notably in frontotemporoparietal and insular regions bilaterally. In the diffusion weighted imaging (D) and the apparent diffusion coefficient map (E), these areas showed marked hyperintensity and hypointensity respectively, characterizing a restriction of water diffusion. Spectroscopy (F) showed a prominent lactate peak (arrow). Since spectral data was obtained at an intermediate echo-time (135 ms), we could not assess glutamine/glutamate peaks, which, if present, would also point to hyperammonemia.

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