META

Glutaric aciduria type I: Ultrasound, computed tomography and magnetic resonance imaging in a child with macrocephaly

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Abstract

[en] The rare case of glutaric aciduria typ I (GA Type I) is described. Its characteristics are discussed and compared with cases in the literature. This disease is basically due to a lack of glutaryl-CoA-dehydrogenase with increased excretion of glutaric acid. Most authors describe frontotemporal cerebral atrophy. In the majority of cases macrocephaly is also present. This sign was also seen in our case and was the reason for performing an ultrasound examination, CT and MR. Ultrasound and CT showed a large insular cistern with incomplete formation of the opercula and frontal atrophy. In addition MR revealed hyperintensity of the basal ganglia and the periventricular white matter. To our knowledge this is the first publication of radiological findings in GA Type I in the German language. (orig.)

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