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BRAIN IMAGING FINDINGS OF METHYLMALONIC ACADEMIA IN 56 CHILDREN

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Background and aims: Methylmalonic acidemia (MMA) is an autosomal- recessive inborn error of organic acid metabolism and caused by an L-methylmalonyl-CoA mutase deficiency. Patients often present with neurological symptoms. We describe the CT and MR brain sectional imaging findings in children with MMA and discuss related pathophysiological mechanisms.

Methods: We retrospectively analyzed CT and MRI of 56 patients images with MMS(41 male,15 female,age range:6 days to 9 years, mean age 11.6months).Brain imaging studies (30MR, 17CT and 9CT+MRI studies) from 56 children were reviewed and reported by two neuroradiologists. The clinical data were collected for each patient.

Results: The most common findings were cortical atrophy (34 studies), ventricular dilation (18), periventricular white matter abnormality (18), subcortical white matter abnormality (18), myelination delay (14), thinning of the corpus callosum (9), cerebellar atrophy (5), basal ganglionic abnormalities (5), increased arterial stiffness and few branches(7). Diffusion-weighted abnormalities were seen in 11 patients with MMA during an acute episode of metabolic acidosis. The brain images in 2 patients were normal.

Conclusions: Radiological findings of MMA are nonspecific. A constellation of common clinical and radiological findings should raise the suspicion of MMA.