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Case Report

MR Brain Imaging of Fucosidosis Type I

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Summary: Fucosidosis is a rare autosomal recessive lysosomal storage disease with the main clinical findings of progressive neuromotor deterioration, seizures, coarse facial features, dysostosis multiplex, angiokeratoma corporis diffusum, visceromegaly, recurrent respiratory infections, and growth retardation. Fucosidosis type I rapidly evolves toward a progressive neurologic deterioration and death. We report MR imaging findings of the brain of three patients with fucosidosis type I, including previously unreported findings, to expand the knowledge of the neuroradiologic spectrum of the disease.

Fucosidosis is a rare autosomal recessive lysosomal storage disease caused by the deficiency of lysosomal enzyme α-L-fucosidase, the enzyme that hydrolyzes fucose from glycolipids and glycoprotein. This results in the accumulation of a variety of α-L-fucose-rich storage products in many organs, such as liver, spleen, skin, heart, pancreas, thymus, thyroid, and kidneys, as well as the brain (1–4). The storage material consists largely of glycoasparagines (glycoproteins), and to a lesser extent, oligosaccharides, mucopolysaccharides, and glycolipids (1–6). The locus for α-L-fucosidase has been assigned to chromosome 1 at position 1p34.l–36.1, and is designated FUCA1 (3). Two clinical variants have been described (2–10). Fucosidosis type I is characterized by early mental and motor regression with rapidly progressive neurologic deterioration to a decerebrate state and death in the first decade of life (1–6). The facial appearance may resemble that seen in mucopolysaccharidosis. Cardiomegaly, hepatomegaly, and anhydrosis are commonly seen. Fucosidosis type II has a milder and more prolonged course, and affected patients often reach adulthood (4–6, 10, 11). Clear-cut distinction between the two types may be complicated by the occurrence of both within one family (2–4). The demonstration of the deficiency of α-L-fucosidase in urine, leukocytes, cultured fibroblasts, or other tissues permits a definite diagnosis.

The purpose of this report is to present the MR imaging findings of the brain of three patients with fucosidosis type 1. Some of these findings are similar to those previously described (5–9), whereas others are newly reported, involving the hypothalamus, putamen, and medullary laminae of the globi pallidi.

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The clinical findings of the three patients are summarized in the Table. In all of them, abnormal oligosaccharide urinary levels, and markedly (patient 1) or undetectable (patients 2, and 3) α-L-fucosidase activity on peripheral leukocytes permitted the diagnosis.

Patient 1

MR imaging of the brain at 0.5 T showed extensive, confluent, and symmetrical signal alteration of the cerebellar, cerebral periventricular, lobar, and subcortical white matter as well as of the internal, external, and extreme capsules, which appeared hyperintense on T2-weighted images. Globi pallidi showed high signal intensity on T1-weighted images and low signal intensity on T2-weighted and fluid-attenuated inversion recovery (FLAIR) images. T2-weighted images showed a subtle hyperintensity in the internal medullary laminae of the thalami. Two curvilinear streaks of low signal intensity on T1-weighted images and increased signal intensity on T2-weighted and FLAIR images were evident within the lentiform nucleus bilaterally, corresponding to the lateral and medial medullary laminae of the globi pallidi. Both putamina were hyperintense on T2-weighted and FLAIR images.

Patient 2

MR imaging of the brain at 1.5 T showed extensive, confluent, and symmetrical signal alteration of the corpus medullare; the periventricular, lobar, and subcortical white matter; the internal, external, and extreme capsules; and the internal medullary laminae of the thalami, which appeared hyperintense on T2-weighted images (Fig 1). T1-weighted images showed relatively high-signal globi pallidi with a hypointense streak between medial and lateral segments of the globi pallidi. On T2-weighted images, this curvilinear streak was hyperintense and the globi pallidi presented abnormal low signal intensity. High signal intensity was evident also in the putamina and in the hypothalamus. MR imaging of the thoracolumbar spine showed anterior and posterior vertebral beaking (Fig 1F).

Patient 3

MR imaging of the brain at 1.5 T showed diffuse, confluent, and symmetrical signal alteration of the corpus medullare as

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