Thalamic Hyperdensity: A Previously Unreported Sign of Sandhoff Disease

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Sandhoff disease is one of the three variants of GM2 gangliosidosis, of which Tay-Sachs disease is the best known [1, 2]. Reports of CT findings in Sandhoff disease are sparse. White-matter hyperdensity has been reported [3]. We describe a 1-year-old infant with biochemically proved disease who had increased density in the thalami on unenhanced CT scans.

Case Report

The patient was born at term after an uncomplicated pregnancy, labor, and delivery. The neonatal course was complicated only by mild hyperbilirubinemia, which was treated with phototherapy. When the baby was 6 months old, the mother sought medical attention because the infant was unable to sit unassisted. A CT scan performed at that time was normal. A second evaluation when the baby was 1 year old revealed an infant at the 50th percentile for weight and the 75th percentile for head circumference who did not track objects. cherry-red spots and optic pallor were present. Muscle tone and mass were diminished, with brisk deep-tendon reflexes. The infant did not exhibit responsive behavior or verbalize.

Enzyme studies showed a lack of hexosaminidase A and B activities consistent with Sandhoff disease. A second CT scan (Fig. 1) showed poor differentiation between gray and white matter; diffuse hypodensity of the brain parenchyma; and symmetric, nonehancing, increased density in the thalami. The infant suffered progressive deterioration and died 1 year later. No autopsy was performed.

Discussion

The gangliosidoses are a subgroup of lysosomal storage diseases in which the absence of enzymatic activity causes the accumulation of biochemical intermediates and resultant cell death. The ganglio-

Fig. 1.—Unenhanced CT scan shows poor definition of differentiation between gray matter and white matter and thalamic hyperdensity. No increase in thalamic attenuation occurred after administration of IV contrast material.

REFERENCES