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AJNR Am J Neuroradiol 1989, 10 (5 suppl) S1-S3
http://www.ajnr.org/content/10/5_suppl/S1.citation

Case Reports

MR Imaging of Cavitary Lesions in the Brain with Hurler/Scheie

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A published report of MR findings in the mucopolysaccharidoses has described increased T2 signal from the periventricular white matter plus enlargement of the ventricles and cortical sulci [1]. We recently used MR to study a girl who had one of the mucopolysaccharidoses. Her MR images showed large cavities within the white matter, as well as diffuse white matter changes and ventricular enlargement. These cavities have been described before in the neuropathologic literature, but we believe this is the first time such cavities have been described as shown by MR or other imaging methods.

Methods

MR was done on a Teslacon* superconducting magnet operating at 0.6 T. Coronal T2-weighted images were obtained with a 3000/33, 66, 99, 132/2 (TR/TE/excitations) sequence. T1-weighted imaging was performed in the sagittal plane with an 833/33/2 sequence and in the axial plane with a 533/33/2 sequence.

Case Report

An 8-year-old girl was seen for evaluation for an enlarged head. Her birth had been normal, after a normal pregnancy. She reached her normal milestones mentally and physically for the first 12 months of life. However, her head circumference enlarged rapidly thereafter, reportedly reaching the 90th percentile by 18 months. When she was first seen in our clinic at age 8 years, she had a large head (56 cm, above the 97th percentile for her age) and a small body (weight, 18 kg; height, 102 cm; both below the third percentile for her age). Her facial features were coarse; she had thick eyebrows and some hirsutism. She had a kyphoscoliosis and contractures of her fingers. She also had corneal clouding and a marked conductive-type hearing loss. Her intelligence appeared near normal, but precise testing was somewhat limited by her visual and hearing impairments.

Laboratory tests showed that (1) the leukocyte alpha-L-iduronidase level was below the detectable range and (2) the urinary mucopolysaccharide level was markedly elevated. MR (Fig. 1) showed marked panventricular enlargement, an enlarged sella, and increased signal in the white matter of the cerebral hemispheres on T2-weighted images. In addition, cavitary lesions in the white matter of the corona radiata were seen and were especially prominent in the parietal and occipital regions. The signal intensity of these cavities closely paralleled the signal intensity of CSF in the ventricles (arrows, Figs. 1A–1C).

Discussion

The mucopolysaccharidoses are a group of syndromes generally characterized by physical deformity and mental retardation. Since the initial description by Hunter, several different subtypes of mucopolysaccharidoses have been reported, each one produced by a defect in one of several different enzymes. The most common form (estimated incidence: 1:100,000 births) [2] was initially described by Hurler in 1919. Hurler disease is now known to be caused by a defect in the enzyme alpha-L-iduronidase. Clinically, one sees mental retardation, coarse facial features, corneal clouding,

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Received June 6, 1988; revision requested August 23, 1988; revision received February 17, 1989; accepted March 1, 1989.

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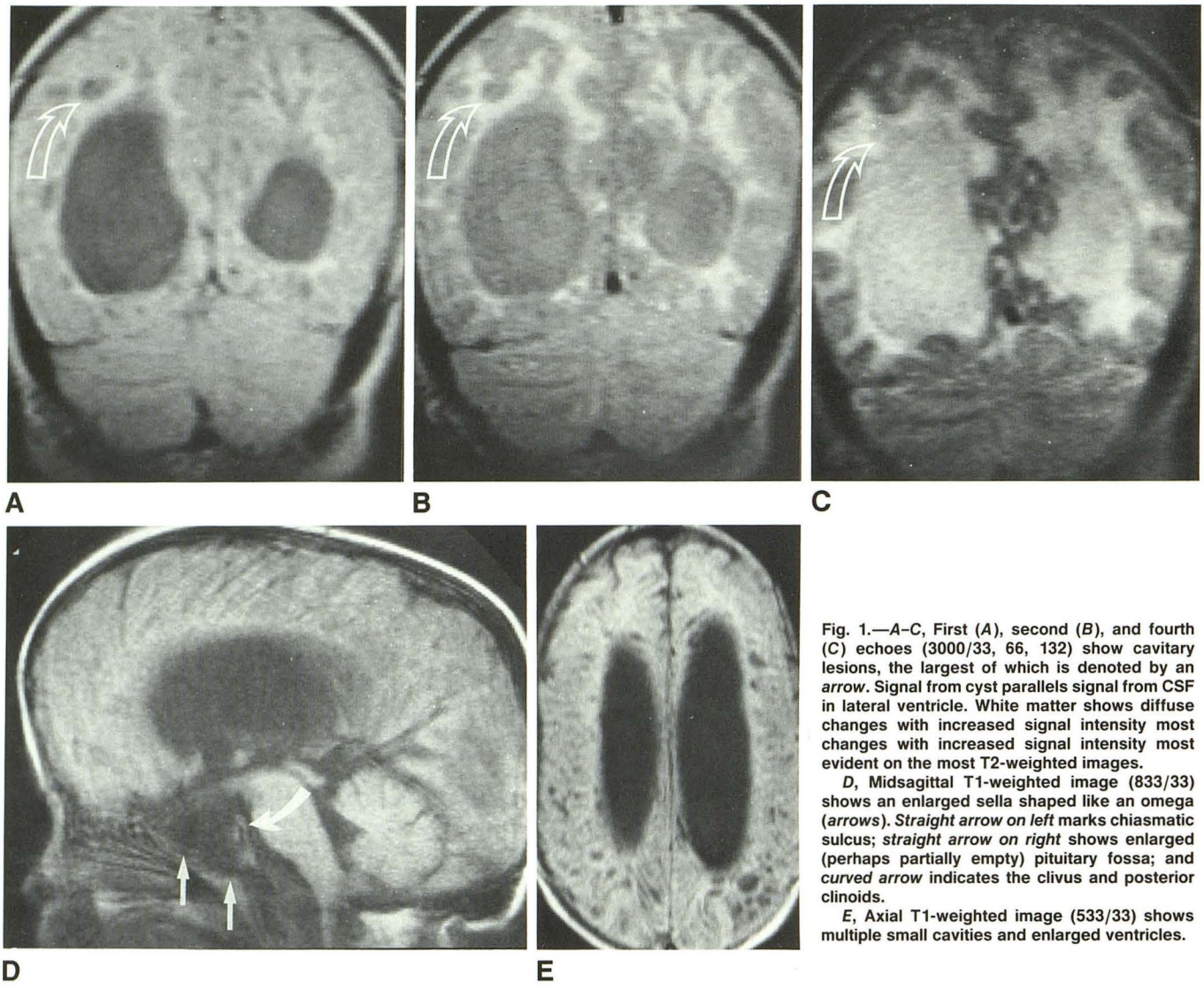

Fig. 1.—*A–C*, First (*A*), second (*B*), and fourth (*C*) echoes (3000/33, 66, 132) show cavitary lesions, the largest of which is denoted by an *arrow*. Signal from cyst parallels signal from CSF in lateral ventricle. White matter shows diffuse changes with increased signal intensity most changes with increased signal intensity most evident on the most T2-weighted images.

***D*, Midsagittal T1-weighted image (833/33) shows an enlarged sella shaped like an omega (*arrows*). *Straight arrow on left* marks chiasmatic sulcus; *straight arrow on right* shows enlarged (perhaps partially empty) pituitary fossa; and *curved arrow* indicates the clivus and posterior clinoids.**

***E*, Axial T1-weighted image (533/33) shows multiple small cavities and enlarged ventricles.**

and death by the early-to-mid teen years. A milder form of Hurler disease was first described by Scheie in 1962. It is now known to be caused by a separate defect in the same enzyme that causes Hurler disease. Both Hurler and Scheie diseases are marked by deafness, short stature, and corneal clouding. The patients with Scheie disease have nearly normal life spans and intelligence. More recently, a Hurler/Scheie genetic compound has been described, in which patients are heterozygous for the two defective genes for alpha-L-iduronidase. These patients have physical findings intermediate between those of Hurler disease and those of Scheie disease [3, 4]. On the basis of clinical findings, we believed that our patient had the Hurler/Scheie compound.

MR in our case showed marked enlargement of all ventricles. Both in the pathologic and radiologic (CT) literature, enlarged ventricles have been reported in many patients with mucopolysaccharidoses [5]. Some of these patients appear to have ventricular enlargement on the basis of diffuse atrophy of brain parenchyma [2]. However, at least some cases are thought to represent obstruction to the flow of CSF by meninges thickened with deposits of mucopolysaccharide, collagen, and fibroblasts [5, 6]. Although our patient has not required a ventricular shunt procedure, the markedly enlarged ventricles with upward bulging of the lateral ventricles and slight depression of the floor of the third ventricle led us to conclude our patient had communicating hydrocephalus.

The sagittal MR images, although somewhat degraded by motion artifact, showed an enlarged sella. In the midline sagittal image (Fig. 1D), the sella appears to be elongated and the chiasmatic sulcus of the planum sphenoidale appears to be abnormally deepened, giving the sella an omega shape. The chiasmatic sulcus is marked by the leftmost straight arrow in Figure 1D. Completing the omega shape are the enlarged (and perhaps partially empty) pituitary fossa (shown by the rightmost straight arrow in Fig. 1D) and the clivus and posterior clinoids (shown by the curved arrow in Fig. 1D). This finding is comparable with the omega- or J-shaped sella described frequently on lateral radiograms of the sella in Hurler disease [7, 8]. Neuhauser et al. [8] postulated that these changes in the shape of the sella were due to arachnoid

cysts around the sella. The T1-weighted sagittal image in our patient did show a signal suggesting fluid in the enlarged sella, which tends to support their hypothesis. However, we cannot say whether this fluid is loculated within an arachnoid cyst or is free within a cistern.

Our patient showed diffuse cerebral white matter abnormality as reflected by high intensity on T2-weighted images (Figs. 1A–1C). In the only other published report on MR in Hurler disease [1], the white matter changes were predominantly in the periventricular white matter. Since those cases also involved hydrocephalus, some of these white matter changes could conceivably have been produced by transependymal reabsorption of CSF. In our patient, almost all the white matter of the cerebral hemispheres was involved. This shows a diffuse abnormality of the patient's white matter; this abnormality can be seen on MR studies. The white matter abnormality is thought to correspond to previous histologic description of gliosis and demyelination in the mucopolysaccharidoses. It also may reflect accumulation of the mucopolysaccharide, both in the perivascular space and (to a lesser extent) in the neurons [2, 9]. In our patient, the white matter of the cerebellum was relatively spared. Similar cerebellar sparing has been reported in the pathologic literature [2]. The mechanism of this sparing is unclear.

The pathologic literature contains many references to small perivascular lacunes or pits visible on the cut surface of the deep white matter in patients with mucopolysaccharidoses [10, 11]. Most of these cysts are perivascular in location and quite small. Usually the largest size of these cysts is only 1 or 2 mm. These small cysts have been reported to contain mononuclear cells as well as a viscous fluid. The mononuclear cells have been called "gargoyle" cells because they contain vacuoles filled with mucopolysaccharide [9]. The pathologic literature also reports a few cases of mucopolysaccharidoses with much larger cavities or cysts (1–6 cm). These have been reported in the corona radiata [12] and the basal ganglia [13]. Our patient had large cavitary or cystlike lesions up to 1.5 cm in diameter in the corona radiata, and we believe this is the first time such cavities have been shown by MR in a patient with a mucopolysaccharidosis.

The pathologists who have previously encountered similar cavities in the brains of patients with mucopolysaccharidoses have wondered about their content. Although several authors have speculated that these cavities may contain mucopolysaccharides [10, 13], the contents of these cavities remain unknown because their contents are altered by fixation before brain-cutting. MR offered a chance to evaluate the cavities in vivo. We had hoped to compare the signal from these cavities with the signals from a CSF collection and a mucopolysaccharide collection. The mucopolysaccharide collection used was the vitreous of the globe, known to contain a large amount of hyaluronic acid, a mucopolysaccharide [14]. However, visual comparison showed no discernible difference between the signal from CSF and that from the vitreous. Thus, visual comparison alone did not allow us to detect mucopolysaccharide within these cavities. We were not able to calculate the T1 and T2 relaxation times for the material in the cavities in our patient. Perhaps if a large enough cavity is found in future research on mucopolysaccharidosis, the T1 and T2 of this cavity will be calculated and compared with the vitreous or other mucopolysaccharide collection. This might allow physicians to determine whether such cavities contain mucopolysaccharide or are simply filled with a CSF-like fluid.

In summary, the MR findings in one patient with Hurler/Scheie disease included ventricular enlargement, enlargement of the sella, diffuse white matter changes, and cavitary lesions. The large cavitary changes are somewhat unusual and have been reported only rarely at autopsy and never in MR studies.

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