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Cranial CT in Fucosidosis

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Fucosidosis is a rare inborn error of metabolism with diminished or absent levels of α -L-fucosidase [1], an enzyme which cleaves fucose from various glycopeptides and oligosaccharides [2]. This results in abnormal storage of fucose-containing substances and abnormal excretion of fucose in the urine [3]. Clinically, there is degeneration of the central nervous system which may progress to spastic quadriplegia and decerebrate rigidity [3].

To date, reports on radiographic abnormalities associated with fucosidosis have been limited to plain-film findings, predominantly skeletal manifestations which are characterized as a mild dysostosis multiplex [4]. We report the cranial computed tomographic (CT) findings in two nonrelated patients with fucosidosis.

Case Reports

Case 1

A 6½-year-old boy was the third child of healthy parents. One brother is healthy, and the other brother died at age 4 of fucosidosis. There is no known consanguinity. The child was apparently normal until 6 months of age, when he was found to be hypotonic. Subsequent evaluation showed coarse facial features with frontal bossing, a widened mouth, short neck with low implantation of the hair, short arms and legs, and coarse hands. Reflexes were normal.

There were radiographic changes of dysostosis multiplex. Urine samples of 24 hr for mucopolysaccharides were negative. Skin biopsy revealed almost total absence of α -L-fucosidase. Initial CT scan of the brain, at age 4, was normal.

The patient has been followed regularly and has had repeated respiratory infections. Spasticity has developed in his legs and language has been severely affected. He has bilateral external strabismus, muscular hypotonia of the paravertebral muscles, increasing kyphoscoliosis, and internal rotation of both feet. Angiokeratoma has developed on the scrotum. Cranial CT at age 6½ revealed normal-sized ventricles but abnormality throughout the corona radiata with focal linear areas of hypodensity, particularly extending up into the gyral surfaces (fig. 1B and 1C).

Case 2

A 17½-year-old boy was the product of an uncomplicated full-term pregnancy. An older sister died at age 4 of fucosidosis. The

patient exhibited normal growth and development until age 2 when he developed a generalized seizure disorder. Electroencephalography at that time was abnormal, showing generalized slow and spiking waves. He subsequently developed speech delay, had slight spasticity in his legs, and had difficulty walking. His face coarsened and his hands became wide and coarse, resembling gargoylism. He continued to show deterioration and, by age 7, was severely mentally retarded, had a kyphoscoliosis with subluxation of his left arm and right hip, and had severe spasticity. He also had generalized angiokeratosis. Skin biopsy for lysozymal enzymes revealed a deficiency of α -L-fucosidase.

By age 17½ his condition had progressively worsened. Cranial CT revealed extreme changes of atrophy (fig. 2). There was dilatation of the ventricular system and marked dilatation of the Sylvian, prepontine, and suprasellar cisterns. Hypodensity was seen along the interhemispheric fissure and over the cortices and in the right cerebellum. The findings are compatible with extreme central, cortical, and cerebellar atrophy.

Discussion

Fucosidosis was first described by Durand et al. [1] as a neurovisceral disease with abnormal accumulation of glycolipid substances in lymphocytes, skin, the liver, the central nervous system, and other tissues. Van Hoof and Hers [5] established that the basic defect was absence or deficiency of the enzyme α -L-fucosidase. This defect leads to the abnormal storage of compounds containing fucose (sphingolipids, oligosaccharides, and polysaccharides) with abnormal excretion of fucose in the urine [3]. Two types are now recognized: type 1, which shows rapid, progressive deterioration and severe neurologic signs, and type 2, in which the neurologic findings are less severe, survival is longer, and skin lesions of angiokeratoma corporis are present [4, 6].

Clinical symptoms become manifest in the first 2 years of life. Patients may have coarse facial features and resemble Hurler syndrome [3]. In addition to the progressive neurologic deterioration, there may be hepatosplenomegaly, cardiomegaly, repeated respiratory infections, thick skin, high concentrations of sweat electrolytes, loss of function of the gallbladder, corneal opacities, angiokeratoma corporis dif-

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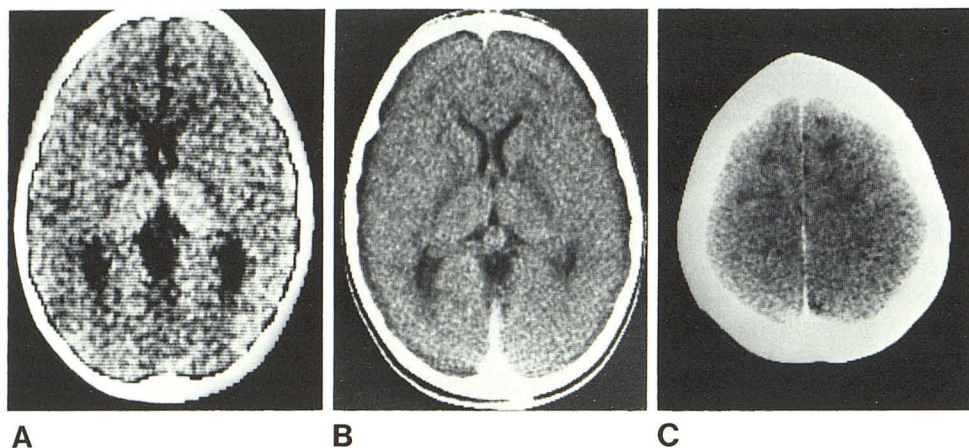


Fig. 1.—Case 1. A, Normal nonenhanced CT (older scanner). B and C, Enhanced CT. Normal-sized ventricles but focal areas of hypodensity.

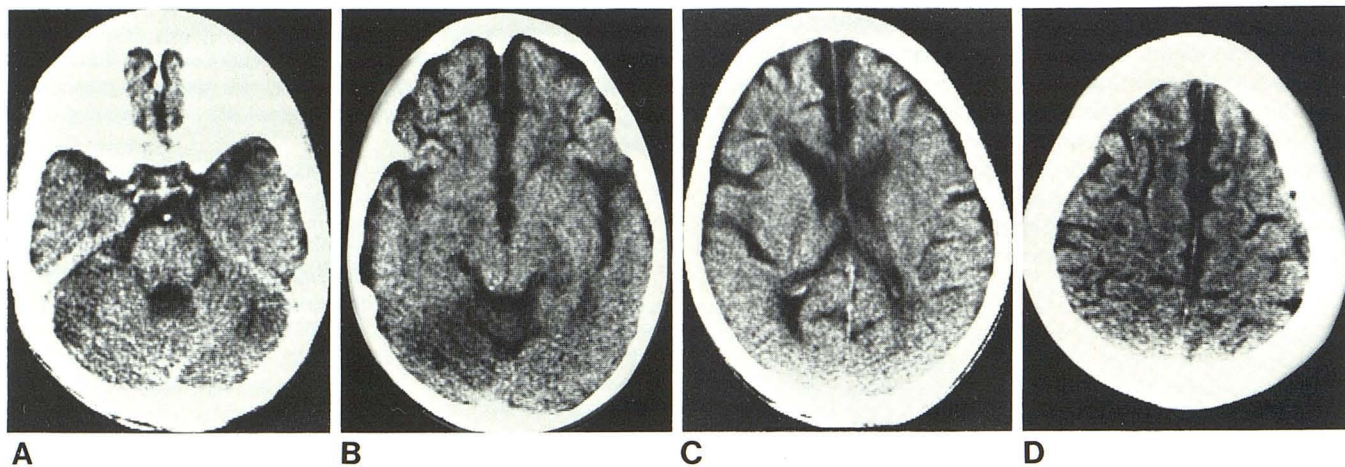


Fig. 2.—Case 2. A and B, Nonenhanced CT. Enlarged fourth ventricle and areas of hypodensity in cerebellum particularly on right. C and D,

Nonenhanced CT. Dilatation of lateral ventricles and cortical atrophy.

fusum, and vacuolized lymphocytes in the peripheral blood. [3].

Neurologically, fucosidosis exhibits progressive neuronal loss with severe demyelination. Microscopic neuropathologic findings in the central nervous system include enlarged nerve cell bodies that are distended and optically empty. Neuronal loss is seen in the cerebral cortex, neostriatum, thalamus, hypothalamus, Purkinje cells, and central nucleus of the cerebellum. Myelination is severely affected and the pathologic picture is one of sudanophilic leukodystrophy [1]. Clinically, there is increasing hypertonia with subsequent spasticity, tremor and, ultimately, decerebrate rigidity. Mental status deteriorates with progressive dementia [1].

The variability of the three CT scans in our patients correlates well with the known clinical progressive nature of the disorder. CT of case 1 at age 4 was normal and, at age 6, showed only mild infiltrative changes in the gray and white matter, whereas that of case 2 at age 17 showed considerable ventricular dilatation associated with cortical and cerebellar atrophy. We suggest that these findings are compatible with progressive central nervous system deteri-

oration. Early demyelination may result in mild changes of gray and white matter, but ultimately marked atrophic changes with concomitant ventricular dilatation will occur.

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