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MR and CT in Cytoplasmically Inherited Striatal Degeneration

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A rare, cytoplasmically inherited striatal degeneration associated with Leber's optic atrophy exhibited selective symmetric low-density lesions in the putamen on the CT scan in five patients. The CT findings, however, were asymmetric (one patient) and subtle in the early phases of the disease. Occasionally, caudate lesions were demonstrable. On MR imaging, the lesions had high signal intensity on T2-weighted images and low signal intensity on T1-weighted images. This group of patients was distinguished from patients with other causes of striatal degeneration by a lack of hemispheric, brainstem, or cerebellar atrophy.

Numerous acute toxic and metabolic encephalopathies as well as a variety of chronic degenerative diseases result in decreased attenuation of the basal ganglia on CT. We report a mitochondrially inherited, progressive striatal degeneration associated with Leber's optic atrophy, which showed similar low-density lesions involving the putamen and caudate nuclei. This condition is further characterized by a distinctive clinical syndrome [1].

Subjects and Methods

Six CT scans of five patients from a large family with a neurologic disorder associated with optic atrophy were studied. These scans were obtained using GE 8800 and GE 9800 CT scanners (General Electric, Milwaukee, WI) with a 10-mm slice thickness. In selected cases, thinner (3-mm) sections were used. The patients ranged in age from 3 to 20 years. One patient was examined twice over a period of 7 years.

One patient was also examined with a GE 1.5-T MR scanner using a partial saturation sequence with TR = 400 msec in axial, sagittal, and coronal planes; and a spin-echo sequence with TR = 2000 msec and TE = 40 and 80 msec in axial and coronal sections.

Results

The pedigree (Fig. 1) of the family revealed that the disorder was transmitted only to the children of maternal lineage, a mode of inheritance known as cytoplasmic or mitochondrial inheritance [2].

The imaging findings and clinical histories of the five patients are summarized in Table 1. The most consistent CT finding was symmetric low attenuation of the putamen (Figs. 2 and 3). Attenuation of the putamen was asymmetric in only one patient. The caudate nucleus was involved in two of the five patients; in one patient this was asymmetric. In one case, thalamic low-attenuation lesions were identified. Despite the long duration of the illness, up to 18 years in one patient (case 1), cortical atrophy was not marked, although prominence of the sylvian fissures relative to the patient's age was seen in two of the five patients.

The putamen nuclei bilaterally had abnormally prolonged relaxation times when compared with other gray-matter structures. Both T1 and T2 relaxation times were

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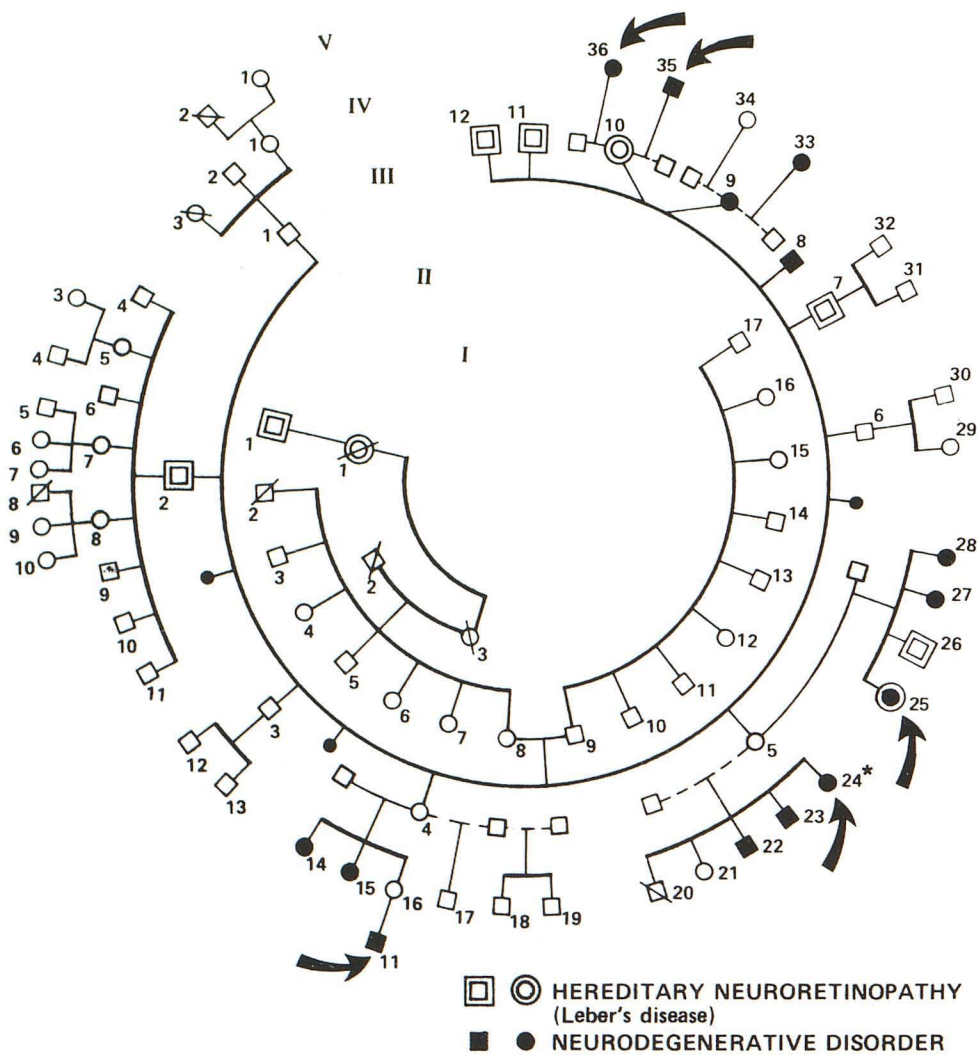


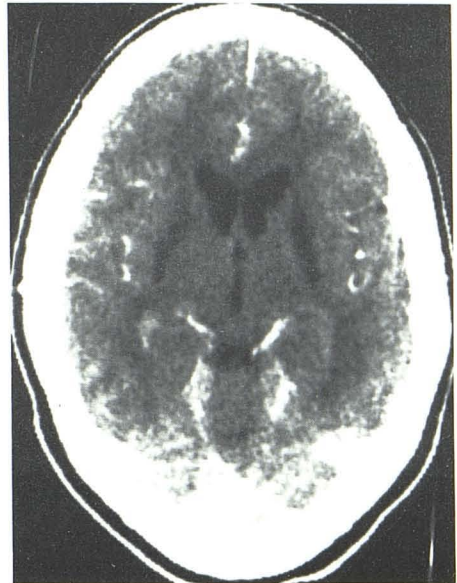
Fig. 1.—Pedigree of family whose CT scans are described. Note maternal inheritance. Squares = male, circles = female, arrows = cranial CT, asterisk = MR.

TABLE 1: Patients and Results of Imaging Studies

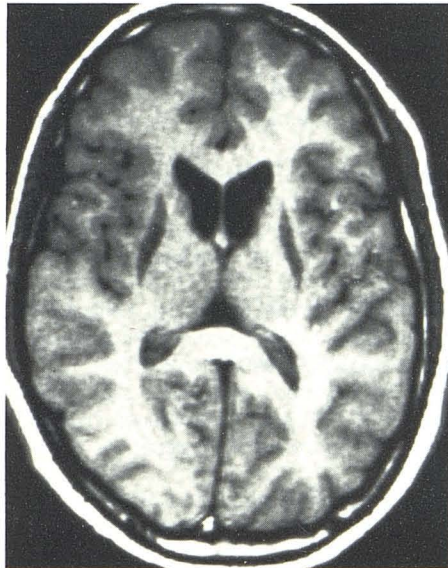
Case No.	Age of Patient When Study Was Done (in years)	Clinical History	Imaging Findings				
			MRI	Putamen (low density)	Thalamic Lesion	Caudate (low density)	Sylvian Prominence
1	18 (CT)	Rigidity and dystonia for 48 months; mental retardation	Elevated T1 and T2 of striatum	Present	Not found	Not found	Present
2	19 (MRI)	Striatal syndrome progressive for 18 months	Not done	Present	Not found	Not found	Not found
	3 (1st CT) 10 (2nd CT)		Not done	Present	Present	Present	Not found
3	4 (CT)	Gait disorder for 18 months	Not done	Present	Not found	Not found	Not found
4	12 (CT)	Pseudobulbar syndrome dystonia; mental retardation for 20 months	Not done	Present	Not found	Not found	Not found
5	16 (CT)	Progressive gait disorder and dystonia for about 18 months	Not done	Present (asymmetric)	Present (L>R)	Present	Present

Note.—For a description of striatal syndrome, see Denny-Brown [17]. L = left; R = right.

Fig. 2.—Case 1. **A**, CT scan showed low density of putaminal nuclei. Caudate lesions were not evident. **B**, Axial MR scan (TR = 400 msec, TE = 25 msec) showed decreased signal intensity of putaminal nuclei bilaterally and of left caudate nucleus. These findings are consistent with a long T1. **C**, Axial MR (TR = 2000 msec, TE = 40 msec) at same anatomical level as **A** and **B** showed increased signal intensity in putaminal and caudate nuclei, indicative of prolonged T2. Caudate lesions were best seen with this pulse sequence. Flow phenomena are thought to account for oval regions of contrasting signal intensity within frontal horns. **D**, Axial MR (TR = 2000 msec, TE = 80 msec) showing prolonged T2 of caudate and putaminal nuclei.



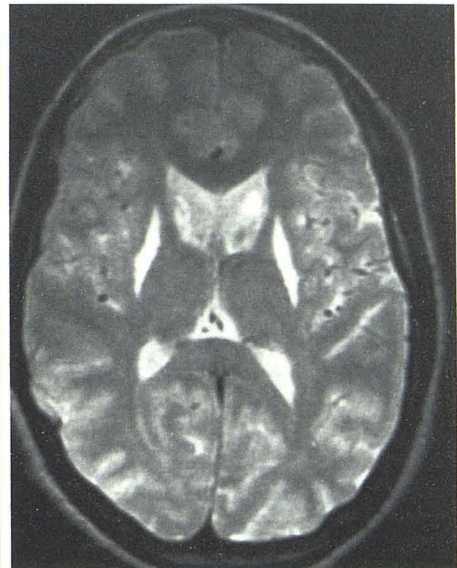
A



B



C



D

prolonged. The caudate nuclei were atrophic bilaterally and showed similar but less marked T1 and T2 changes. The thalami in this patient were normal.

Discussion

Several familial and sporadic neurodegenerative disorders have been described. The condition reported here can be distinguished from them by means of CT and MR findings, the pattern of inheritance, the clinical course, and other laboratory tests. Imaging findings center around the corpus striatum, which shows changes consistent with tissue loss. In one patient, thalamic lesions were present in the centro-median thalamic nucleus, which is known to supply afferent

fibers to the basal ganglia [3]. It is thus reasonable to conclude that a form of retrograde degeneration may have caused this abnormality. Thalamic lesions, however, were not a constant feature.

Several diseases enter the differential diagnosis. Wilson's disease produces symmetric low-attenuation lesions in the putamen, accompanied by cortical, cerebellar, and brainstem atrophy. The globus pallidus may be involved as well [4]. The MR appearance may be similar to that seen in our patient, but atrophy would also be observed [5]. A clinical diagnosis of Wilson's disease is usually made before CT abnormalities appear [6]. Leigh's disease, a recessively inherited nonpyramidal motor disease, can also involve the putamen on CT, but brainstem and cerebellar atrophy are usually present [7–

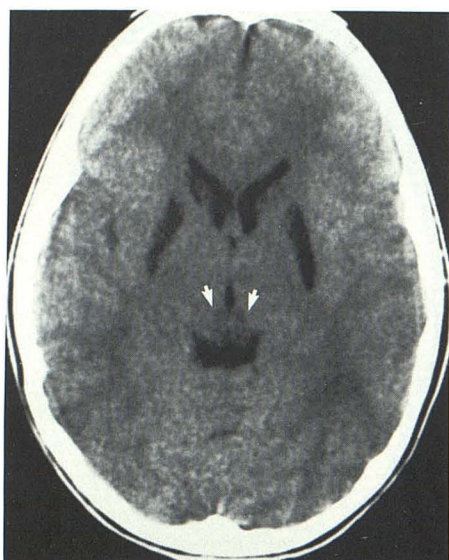


Fig. 3.—Case 2. Axial CT section through basal ganglia showed both caudate and putamen low density. Sylvian fissures were minimally prominent. Low density of left centromedian nuclei of thalamus (arrows) was detected. Concentric rings of alternating density in left thalamus are result of detector calibration artifact.

9]. MR findings have not been reported in this condition. Hallervorden-Spatz disease presents in early childhood, as seen in our patients, but it is relentlessly progressive and can cause focal caudate atrophy (demonstrable on CT) [10]. Johnson et al. [5] describe long T1 and T2 relaxation times in the putamen without obvious cortical atrophy. The report by Littrup and Gebarski [11] is somewhat less clear, but lesions had a long T1 judging from their intensity compared with that of brain and cerebrospinal fluid. The nosology of this disease is complex, making diagnostic error a possibility. It may be that different stages of this disease display different abnormalities. Despite imaging similarities, the hereditary pattern of the syndrome reported here is distinct from that of Hallervorden-Spatz disease [1].

Disorders associated with mitochondrial disease often produce basal ganglia lucency on CT. Egger and Kendall [12] described low density of the lentiform nuclei in the "ophthalmoplegia plus" syndrome, which is also termed mitochondrial cytopathy or Kearn-Sayre's syndrome. In this syndrome, marked cerebral hemispheric white-matter low density has been observed, a finding not present in our patients [13, 14]. These patients can also be distinguished from our patients on clinical, genetic, and pathologic grounds. A single, apparently sporadic case of progressive striatal degeneration and optic atrophy with CT findings similar to ours has been described previously in a patient who was clinically diagnosed as having Leigh's disease [15]. Nevertheless, a sporadic case of a disorder similar to the one reported here is a possibility as well.

The clinical condition described here has been reported previously as "motor degeneration" [16]. The fact that only

affected females in the current family can pass on the disease provides strong evidence that cytoplasmic inheritance rather than sex-linked transmission is occurring [1]. It is hypothesized that nonnuclear genetic material, presumably mitochondrial DNA, is defective in disorders transmitted in this manner [2]. Mitochondrial or cytoplasmic abnormalities are presumably present in each of the patient's cells, so clinical manifestation primarily in the basal ganglia is of interest. We are unaware of any explanation for this phenomenon. It is also curious that the caudate and putamen, embryologically continuous with one another and indistinguishable in some species, are apparently differentially affected.

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