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Adrenoleukodystrophy: New CT Findings

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Three cases of adrenoleukodystrophy are presented with sequential findings by computed tomography (CT). In two cases, CT confirmed not only the findings of several previous reports of a caudocranial direction of low-density white matter and contrast enhancement at the periphery of affected regions during the active demyelinating process, but also two other unusual features: (1) calcification in the affected white matter along the trigones of the lateral ventricles and (2) development of a mass effect during the active demyelinating period. Calcification alone without low-density white matter was the initial CT presentation in one case. Periventricular calcification may be specific for adrenoleukodystrophy in the appropriate clinical setting.

Although brain biopsy is essential for the diagnosis of adrenoleukodystrophy, age, gender, characteristic clinical manifestations, and poor response to adrenocorticotrophic hormone are virtually diagnostic. The most helpful radiologic examinations are computed tomography (CT) and radionuclide brain scintigraphy [1-6]. CT scans may appear normal in the early stage, but show low density involving the white matter or atrophic change sometime in the course of the disease. Radionuclide scintigraphy is occasionally helpful, reflecting its abnormal distribution where the disease is active [3, 5, 6].

Since installation of a CT scanner in 1977, we have encountered cases of adrenoleukodystrophy, each of which demonstrated calcification in the occipitoparietal white matter around each lateral ventricle. We are only aware of one other recent similar case report in the English literature [7]. Since this has not been reported in any other leukodystrophy, calcification of this type might be diagnostic for adrenoleukodystrophy.

Case reports

Case 1

A 5½-year-old boy was evaluated for progressive paraparesis and hypertonia of 1 year duration. There was no history of meningoencephalitis. Family history revealed that his cousin had died of adrenoleukodystrophy at age 10 years, which was confirmed by autopsy. His cerebrospinal fluid (CSF) protein was 50 mg/dl and an electroencephalogram (EEG) was abnormal but not specific. A CT scan at age 5 years was normal, as was cerebral angiography.

Over the next 6 months, he developed intellectual dysfunction and an expressionless face. CT (figs. 1A-1C) at 5½ years revealed mild ventricular and sylvian fissure dilatation and abnormal low-density areas in the frontal and parietal white matter bilaterally. Calcification was demonstrated in the periventricular white matter in the parietooccipital lobes. Postcontrast scans showed slight enhancement along the occipital horns. The patient's condition continued to deteriorate with the development of decorticate posture and generalized seizures, which were not well controlled by anticonvulsants.

Complete blood count, urinalysis, serum amino acids, and lysosomal enzyme activity were normal. CSF protein was normal at admission, elevated (106 mg/dl) at age 5¾ years,

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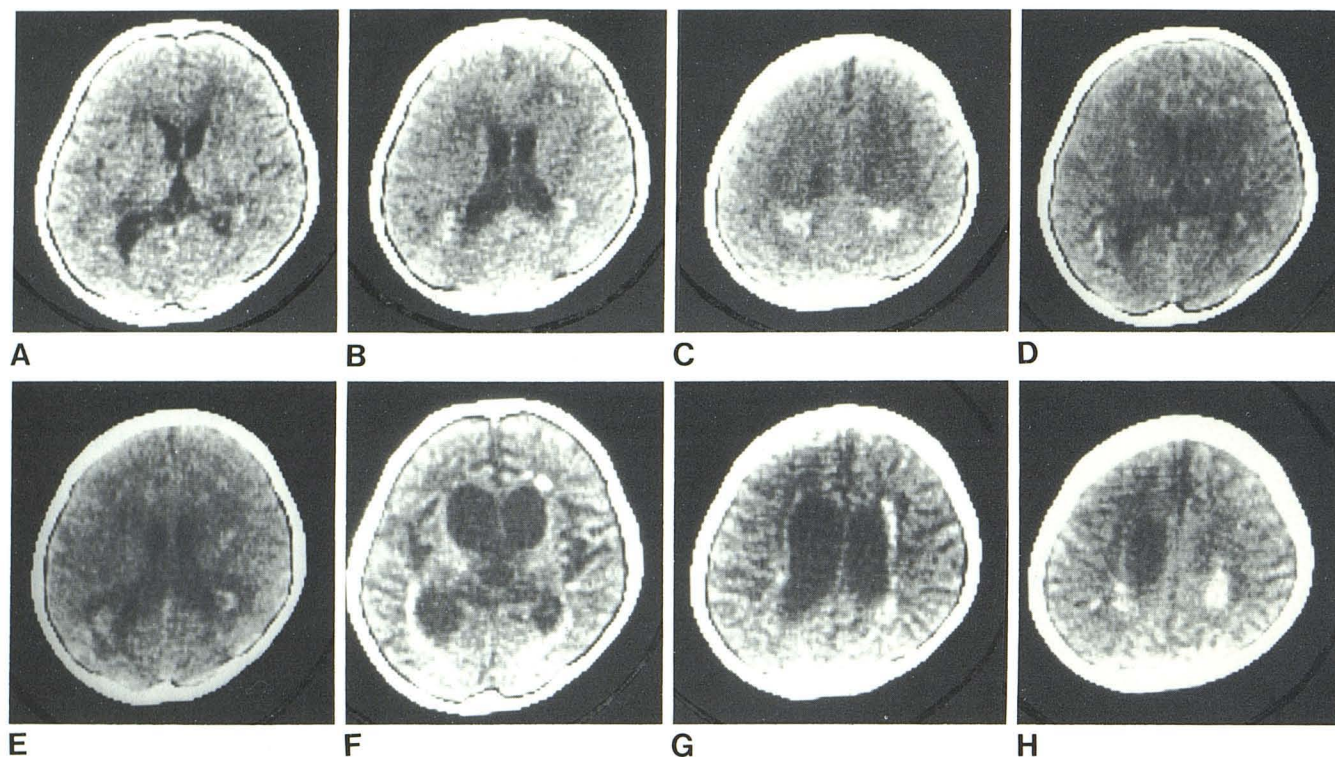


Fig. 1.—Case 1. Precontrast CT studies. A–C, At 5½ years of age. Minimal ventricular dilatation, abnormal low density in both frontal and parietal white matter, and calcification adjacent to the occipital horns. Postcontrast scans showed slight enhancement near calcified areas. D and E, At 5¾ years old. Compressed small ventricle and progressive low density in white matter

of parietooccipital lobes. Calcification appears to be enlarging. New calcified deposits in frontal lobes. F–H, Moderate ventricular dilatation and further periventricular calcification. Abnormal low density in white matter is less prominent. Sylvian fissure and cerebral sulci are also dilated. No abnormal enhancement was seen after contrast injection.

and back to normal 4 months later. Serum ACTH was also normal at admission, but increased at just over 6 years of age to 224.9 pg/ml (normal, 20–100 pg/ml), and remained elevated thereafter. Serum cortisol response to ACTH stimulation was normal at the beginning of the disease but became reduced inversely to the increased serum ACTH value.

Although a brain biopsy was not performed, the diagnosis of adrenoleukodystrophy was suggested by the findings of adrenal insufficiency in a young male with signs of progressive, diffuse central nervous system dysfunction and white-matter low density visualized on CT scans.

The CT scans at 5¾ years (figs. 1D and 1E) demonstrated compressed small ventricles and progression of the low-density white matter to involve the parietal and occipital lobes. The occipital horn calcification appeared to enlarge, and new calcification developed in the frontal white matter. Postcontrast CT showed abnormal enhancement at the edge of the involved white matter.

CT at age 6½ years (figs. 1F–1H) revealed moderate ventricular dilatation with less prominence of the abnormal white-matter low density. Further calcification was noted in the frontal and temporal lobes. No abnormal enhancement was demonstrated after contrast injection.

Case 2

A 4½-year-old boy was admitted after 3 months of decreased visual acuity and unsteady gait. There was no history of meningoencephalitis. In addition to visual disturbance, examination revealed bilateral hyperreflexia and bilateral Babinski reflexes. Complete

blood count, urinalysis, urine organic acids, CSF analysis, and lysosomal enzymes were normal. EEG was abnormal but nonspecific. Skull films were normal. Precontrast CT showed low-density white matter in both temporooccipital lobes and irregular-shaped calcification in the white matter along the posterior horn of each lateral ventricle. There was mild, ringlike contrast enhancement at the edge of the low-density area. The diagnosis of adrenoleukodystrophy was suggested. A radionuclide brain scan showed bilateral symmetric uptake in the parietooccipital region characteristic of adrenoleukodystrophy. Serum ACTH was increased to 200 pg/ml, and the serum cortisol response to ACTH stimulation was poor.

Hypertonia and paraparesis developed soon after admission. Over the next several months, the patient's condition slowly deteriorated, resulting in a decorticate posture and intellectual impairment. The second CT scan (fig. 2) 2 months after admission revealed a slightly increased amount of calcification in both parietooccipital lobes. No enhancement was seen after contrast injection.

Case 3

A 4-year-old boy developed fatigue, appetite loss, and intermittent vomiting followed by cutaneous pigmentation. Addison disease was diagnosed, and corticosteroid treatment was started. There was no history of meningoencephalitis. At age 6 years he developed psychotic changes and visual disturbances, resulting in complete blindness in a short period. CT at age of 7½ years showed calcification in the parietooccipital white matter along the posterior part of the lateral ventricle bilaterally. No white-matter low density was demonstrated.

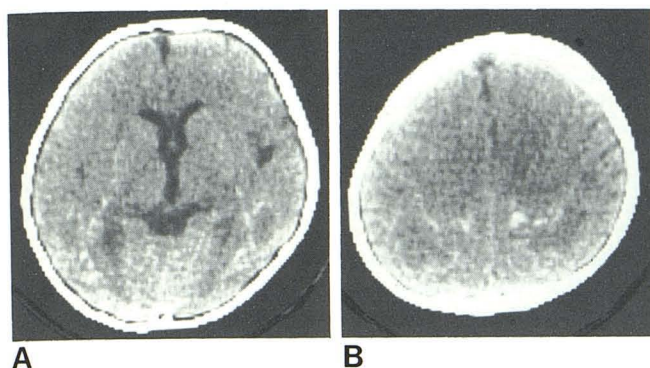


Fig. 2.—Case 2. Precontrast CT scans at 4½ years of age. Small calcification in white matter of both parietooccipital lobes. No abnormal enhancement was observed on postcontrast studies.

Progressive neurologic deterioration was noted. Serum ACTH was elevated to 740 pg/ml, and adrenoleukodystrophy was diagnosed. A second CT study at age 7½ years revealed increased periventricular calcification and localized low-density areas in the parietooccipital white matter just posterior to the calcification (fig. 3). No abnormal enhancement was noted after contrast injection.

Discussion

Adrenoleukodystrophy is a primary demyelinating disease of childhood characterized by widespread demyelination of the cerebral white matter, starting occipitally and spreading frontally, sparing the subcortical U fibers. The clinical and pathologic features of the disease have been comprehensively reviewed by Schaumburg et al. [8], who found adrenal lesions at autopsy in most of the cases, whether or not the patients had adrenal insufficiency during life. The disease presents in boys of school age with cortical visual loss, intellectual apathy, and an atactic-spastic gait. Spasticity progresses until the patient lies in a decorticate state. Seizures occur later. Although the genetic basis of the disease is unclear, the studies of Suzuki et al. [9] and Igarashi et al. [10, 11] have revealed the presence of very long-chain fatty acids in the brain and adrenal glands. Although we had no pathologic proof in our three cases, the clinical, biochemical, and CT appearances were very suggestive of adrenoleukodystrophy.

CT is a promising diagnostic technique in the appraisal of adrenoleukodystrophy. The CT findings described previously [1–4] are: (1) low-density white matter around the trigone of each lateral ventricle, with subsequent involvement of the temporo-frontal lobe white matter (craniocaudal direction), and (2) contrast enhancement at the periphery of the affected regions, although this is an inconsistent feature.

Our present work describes two unusual CT features of adrenoleukodystrophy. The first unusual feature, which confirms a previous observation, is calcification in the involved white matter, especially around the trigones of the lateral ventricles, as seen in cases 1 and 2. Of interest in case 3, the periventricular calcification around the trigone was initially the only abnormal finding, with no evidence of low-density areas in the white matter.

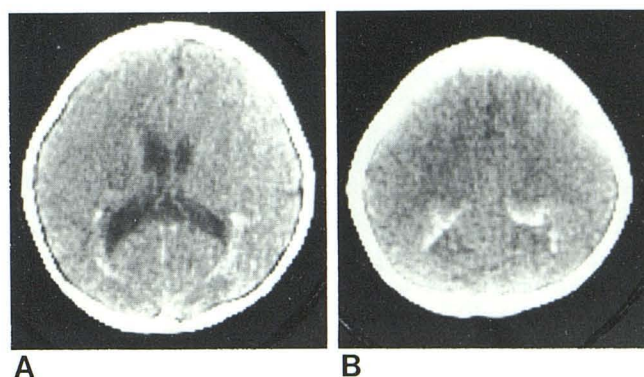


Fig. 3.—Case 3. Precontrast CT scans at age 7½ years. Periventricular calcification in parietooccipital region. Localized low-density areas in white matter just posterior to calcification. No abnormal enhancement was seen on postcontrast studies.

While intracerebral calcification can be produced by many other causes, the absence of a history of meningoencephalitis excluded postencephalitic calcification. Drug-induced calcification was unlikely because no specific drugs were administered except in case 3, in whom corticosteroids were given for 3 years. Intracerebral calcification in adrenoleukodystrophy was described by Schaumburg et al. [8]. They found small deposits of calcium in two of 17 autopsy brains with adrenoleukodystrophy in a zone of gliosis involving the parietooccipital white matter, which corresponds topographically to the low-density area in which calcification was detected on CT in our cases. Therefore, calcification in the parietooccipital white matter along the trigones of the lateral ventricles may be specific for adrenoleukodystrophy in the appropriate clinical setting [7]. The etiology of the calcification in the involved white matter is unclear, although degeneration or necrosis is generally proposed as a possible cause.

The second unusual feature is the demonstration of a mass effect during the active period of adrenoleukodystrophy (case 1). It is well known that the abnormal low density in the white matter is present for only a limited time, corresponding to active dys- or demyelination, and is followed by severe atrophy [2, 12]. However, a mass effect in adrenoleukodystrophy has not, to the best of our knowledge, been reported previously. Although the pathogenesis of a mass effect in leukodystrophy is not well understood, an accumulation of water during the early, active degradation period in white-matter disease [13] seems to play an important role.

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Editor's Note

See also the article: Aubourg D, Diebler C. Adrenoleukodystrophy—its diverse CT appearances and an evolutive or phenotypic variant: the leukodystrophy without adrenal insufficiency. *Neuroradiology* **1982**;24:33-42