Fahr Syndrome

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A 57-year-old man presented with an insidious onset of balance difficulties, including dizziness and vertigo, for some time. The patient's symptoms had progressed to the point at which he had to hold onto things to get around the house. He had developed upper- and lower-extremity dexterity problems. Over time, he had become slow with nearly all of his daily activities and had developed dysarthria. As the man's condition progressed, he had begun to experience tremors in the upper and lower extremities, and expressive aphasia, including cognitive impairment.

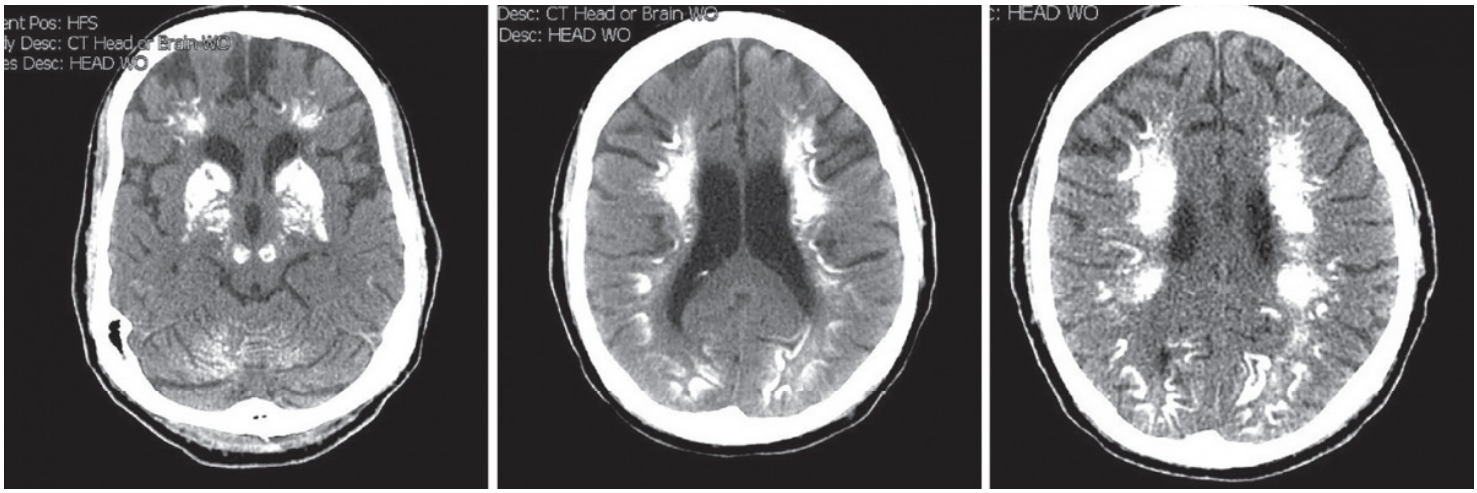

Figure. *Noncontrast CT images demonstrating extensive multiple calcification in the brain parenchyma, cerebellum, basal ganglia, thalamus, frontal, temporal, and occipital white matter.*

Physical examination. The initial physical examination revealed stable vital signs, including a blood pressure of 110/70 mm Hg, a pulse of 80 beats/min and regular, a respiratory rate of 18 breaths/min and regular, and a temperature of 37.1°C. There were no carotid bruits, no cardiac murmurs, no hepatosplenomegaly, and no edema of the extremities. His neurologic condition had changed from bradykinesia, resting tremors, and hyperreflexia to quadriplegia and areflexia.

Diagnostic tests. Electrocardiography findings and levels of calcium, phosphorus, alkaline phosphatase, and albumin were normal. A noncontrast computed tomography (CT) scan of the head demonstrated extensive calcifications involving the bilateral basal ganglia, caudate nuclei, thalami, red nuclei, and dentate nuclei (**Figure**). The pattern and distribution of the calcifications were nonspecific but compatible with Fahr disease, especially given the man's clinical presentation.

Discussion. Fahr syndrome, also known as idiopathic basal ganglia calcification or bilateral striopallidodentate calcinosis, is a rare, genetically dominant, inherited, degenerative, neuropsychiatric disorder characterized by extrapyramidal and bilateral vascular calcification of the basal ganglia.[1,2]

The condition is named for German neurologist Karl T. Fahr, who noted idiopathic calcification in the cerebral vessels of a patient in 1930.[3] Clinical symptoms include seizures and deterioration of speech and motor functions. The typical age at the onset of clinical symptoms is from 30 to 60 years; however, unusually early onset has been reported.[1,4]

The prevalence of Fahr syndrome is not known. Approximately 0.7% of noncontrast CT scans show some calcification of the basal ganglia as an incidental finding that usually is benign, especially in patients older than 60

years.[5] There are 2 forms of Fahr syndrome according to the etiology of the disease.[6] The primary form is genetic or familial, while the secondary form is caused by disorders such as hypoparathyroidism, hyperparathyroidism, or pseudohypoparathyroidism.[7]

The exact cause of Fahr syndrome still is unknown and is a subject of research. By performing a whole-genome scan of 24 members of a family, researchers have identified the first chromosomal locus (14q) for this disorder.[5] Subsequently, second[8] and third[9] loci have also been reported on chromosome 8 and chromosome 2, respectively, indicating genetic heterogeneity.[10]

There is no known cure for Fahr syndrome, and treatment is tailored to address the symptoms on an individual basis.

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