Transcranial Sonography of Basal Ganglia Calcifications in Fahr Disease

Fahr disease, also known as bilateral striopallidodentate calcinosis, is a rare degenerative neurologic disorder characterized by almost symmetric calcifications of the basal ganglia, the cerebellum dentate nuclei, the thalami, and the white matter of the cerebral hemispheres. Bilateral striopallidodentate calcinosis manifests as familial autosomal dominant but also as sporadic forms. Movement disorders (both Parkinsonism rigidity and hyperkinesia) represent the most common clinical manifestations of Fahr disease, usually followed by cognitive and cerebellar impairment.

Neuroimaging techniques, such as cerebral computed tomography (CT) and magnetic resonance imaging (MRI), when correlated with a typical clinical history and sometimes with phosphocalcic assessment (ie, hypocalcaemia and hyperphosphorhemia in cases of bilateral striopallidodentate calcinosis secondary to hypoparathyroidism), are highly suggestive of bilateral striopallidodentate calcinosis: typical bilateral intracranial calcinosis can be identified. Recently, transcranial sonography has been recognized as a reliable and sensitive technique for the detection of basal ganglia abnormalities in several movement disorders, such as Parkinsonism, corticobasal degeneration, Wilson disease, and other extrapyramidal disorders. To our knowledge, no cases of basal ganglia alterations in Fahr syndrome as shown with this technology have been reported previously.

A 54-year-old man was admitted to our neurologic ward for a sudden worsening of extrapyramidal symptoms with gait freezing and postural instability. His clinical history started several years before, and he was receiving L-3,4-dihydroyxphenylalanine treatment. Cerebral MRI at admission showed extensive calcifications involving the basal ganglia, dentate nuclei, and cerebellar hemispheres, supporting Fahr syndrome (Figure 1). Transcranial sonography was performed through the temporal acoustic bone windows using a phased array ultrasound system equipped with a 2-MHz transducer (S2000; Siemens AG, Erlangen, Germany). The sonographic parameters were set according to previous criteria indicated in the literature: the insonation depth was set at 13 cm to the visualization of the opposite temporal bone; the dynamic range of the images was set at 50 dB; persistence was set on high; and reject was set on 7. The image brightness and time gain compensation were adjusted to the best contrast. The brain stem, basal ganglia, and ventricles were investigated on standardized axial transcranial sonographic scanning planes. We observed increased echogenicity in the substantia nigra in the mesencephalon (Figure 1A), diencephalon (Figure 1B), thalamus (Figure 1C), and lateral ventricles (Figure 1D). All of these findings corresponded to MRI abnormalities (Figure 1, right panels).

Neuroimaging techniques play a pivotal role in the diagnosis of bilateral striopallidodentate calcinosis because both CT and MRI are able to show intracranial calcifications. To date, CT remains the most effective screening tool in defining the site and extent of the calcifications, but false-negative results may still arise because the minimum age at which negative CT findings exclude the disease in not established. Magnetic resonance imaging seems to be more sensitive in depicting the various stages of the disease because low–signal intensity areas on T2-weighted spin echo sequences reflect an early stage of calcium deposition, whereas high-signal intensity areas probably reflect a later stage of calcinosis, with increased proteins and endothelial membrane incompetence. Nonetheless, a potential limit of MRI for Fahr disease is that images of calcifications may show different signal intensities, thus confounding its differential diagnosis.

Transcranial sonography is a very diffuse, relatively low-cost, noninvasive imaging technique that finds application not only in cerebrovascular diseases but also in several other neurologic conditions, such as brain parenchymal imaging in movement disorders, when the temporal bone window is adequate for the investigation. Transcranial sonography can indeed reveal basal ganglia degeneration and altered echogenicity of the substantia nigra even in early Parkinson disease but also in other degenerative disorders such as Wilson disease. To our knowledge, a well-documented report of transcranial sonography in Fahr disease in which calcifications of the basal ganglia (which cause hyperechogenicity on sonography) could be easily identified has not been published previously. This case further confirms the utility of transcranial sonography in the evaluation of brain parenchymal changes in movement disorders.

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References


Figure 1. Basal ganglia calcifications in a 54-year-old man with Fahr disease: B-mode sonograms of brain parenchyma through different axial planes (A, mesencephalon; B, diencephalon; C, thalamus; and D, lateral ventricles) showing hyperechoic spots related to basal ganglia calcifications (arrows). Note the corresponding alterations in the fast field echo magnetic resonance imaging sequences (right panels).