

Symmetric Calcifications of the Basal Ganglia and Thalamus: About a Case of Fahr Disease

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Clinical History

A 30-year-old man was admitted in our Neurology department for one-month standing paresthesia of his left hemi-body and a left-hand disability.

His personal and familial medical history was unremarkable, except benign paroxysmal positional vertigo. The clinical examination was normal. Blood and cerebrospinal fluid analysis were normal. Somatosensory evoked potentials did not show any relevant anomaly. Cerebral CT and MRI were performed.

Imaging findings

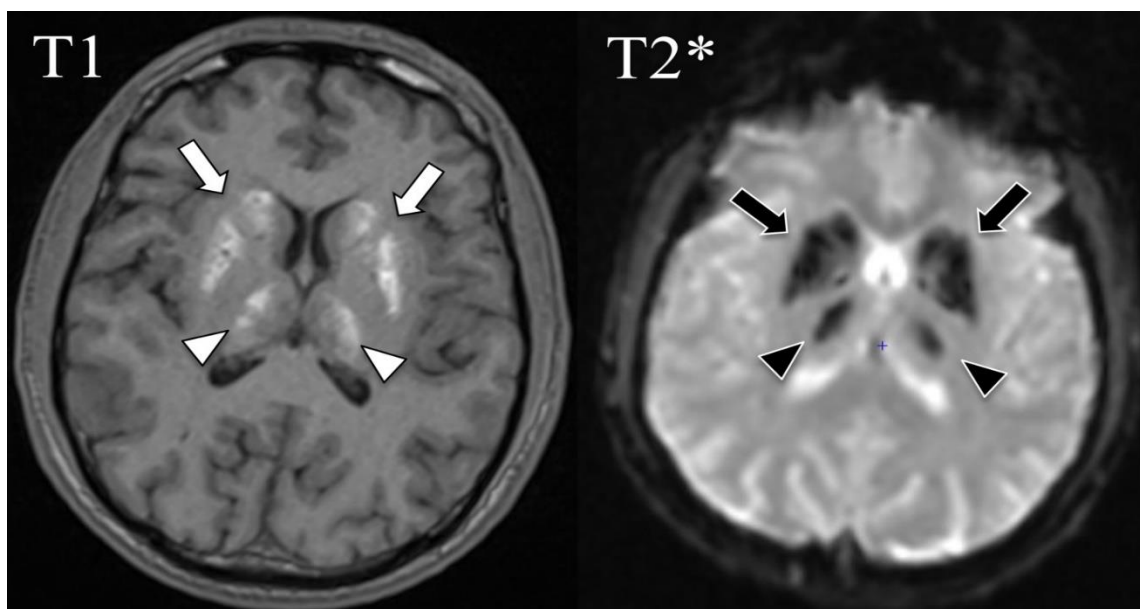


Figure A: Axial brain MR views showing a high intensity signal of the thalami (white arrow heads), lentiform and caudate nuclei (white arrow) on T1-weighted images, and a very low intensity signal in T2*-weighted images (black arrows and arrow heads), highly suggestive of calcifications.

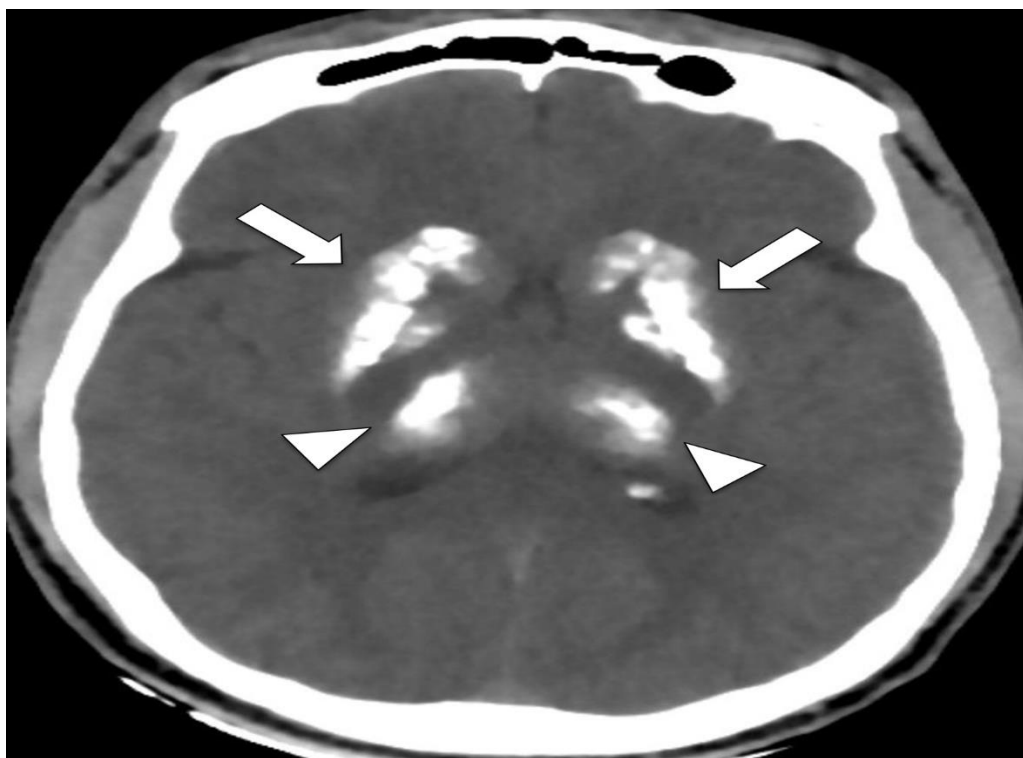


Figure B: Axial brain CT view showing symmetric diffuse calcification of the basal ganglia (white arrows) and of the thalamus (white arrow heads).

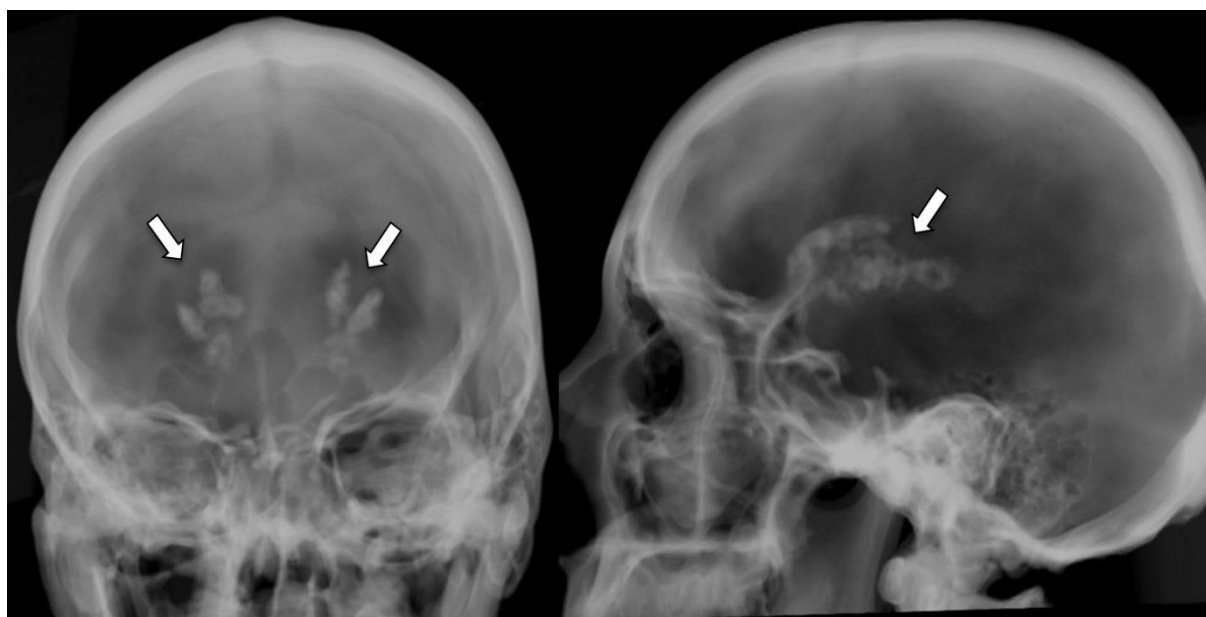


Figure C: Anteroposterior and lateral thin multiplanar reconstruction CT views (mimicking skull radiographs) demonstrating deep intracranial calcifications (white arrows), in the area of the basal ganglia and thalamus. Those images demonstrate that such calcifications could be seen fortuitously on skull radiographs.

Diagnosis

Based on the radiological findings and the clinical story, the diagnosis of **Fahr disease** was suspected.

Discussion

Fahr disease, also called idiopathic basal ganglia calcifications, is a rare neurodegenerative disease, characterized by bilaterally calcification of the basal ganglia, thalamus, dentate nuclei and centrum semiovale. Both familial (autosomal-dominant fashion in most of case) and non-familial cases have been described.

The clinical expression is not specific and numerous signs and symptoms are associated with this disease: neurological features (e.g. loss of consciousness, tetany, seizures, epileptic disorder, gait disturbance, vertigo, headache), movement disorders (e.g. fatigability, involuntary movement and muscle cramping) and neuropsychiatric disorders (psychoses, depression...). The disease is yet incurable and treatment is symptomatic.

The deep grey matter nuclei are composed by basal ganglia and thalamus. The thalamus is a relaying sensory and motor center and regulates the consciousness, sleep and alertness. The basal ganglia are involved in the extrapyramidal motor system but also in memory, emotion and cognitive functions. They are rich in mitochondria and their high metabolic activity make them vulnerable to numerous metabolic, systemic and generalized dysfunctions. Thus, the differential diagnosis of bilateral anomalies of basal ganglia and thalamus is extensive (intoxications, neoplasia and inflammatory/infectious, vascular, metabolic and degenerative disease).

The diagnostic criteria included:

- Bilateral calcification of the basal ganglia visualized on neuroimaging. Other brain regions may also be observed.
- Progressive neurologic dysfunction. Age of onset is

typically in the fourth or fifth decade.

- Absence of biochemical abnormalities and somatic features suggestive of a mitochondrial or metabolic disease or other systemic disorder.
- Absence of an infectious, toxic, or traumatic cause.
- Family history consistent with autosomal dominant inheritance.

The differential diagnosis is wide and must include phosphocalcic metabolism anomalies (principally (pseudo)hypoparathyroidism), mitochondrial disease, genetic syndromes (personal and familial history must be carefully checked) and other causes of intra-axial calcifications such as vascular, infectious, neoplastic and toxic diseases.

Bilateral calcification of the basal ganglia is a really common finding in head imaging: CT, MRI but also skull radiography (as demonstrated on Fig.C, profuse calcifications can be an incidental finding when skull radiographs are performed, e.g. for sinus visualisation, myeloma monitoring, traumatic indications or to detect ferromagnetic foreign bodies before MRI examination). Fahr disease must be suspected when the imaging fit with the medical history and clinical findings we described earlier.

Competing Interest: The authors declare that they have no competing interests.

Reference

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