

Incidental Fahr's disease

Incidentální Fahrova nemoc

Dear Editor,

Fahr's disease, also known as bilateral symmetric striopallidodentate calcinosis, is a rare neurodegenerative disorder characterized by symmetric intracranial calcifications, most often involving the basal ganglia, thalamus, dentate nuclei, and cortical regions. It is usually inherited in an autosomal dominant manner, although sporadic and autosomal recessive cases have been described [1]. Mutations in the *SLC20A2* gene, encoding the sodium-dependent phosphate transporter 2, are considered the most frequent genetic cause. The condition must be differentiated from Fahr's syndrome, in which similar calcifications develop secondary to metabolic or systemic causes, most notably hypoparathyroidism or pseudohypoparathyroidism [2]. Clinical presentation is heterogeneous, ranging from extrapyramidal symptoms such as dystonia, tremor, chorea, and parkinsonism to cognitive and psychiat-

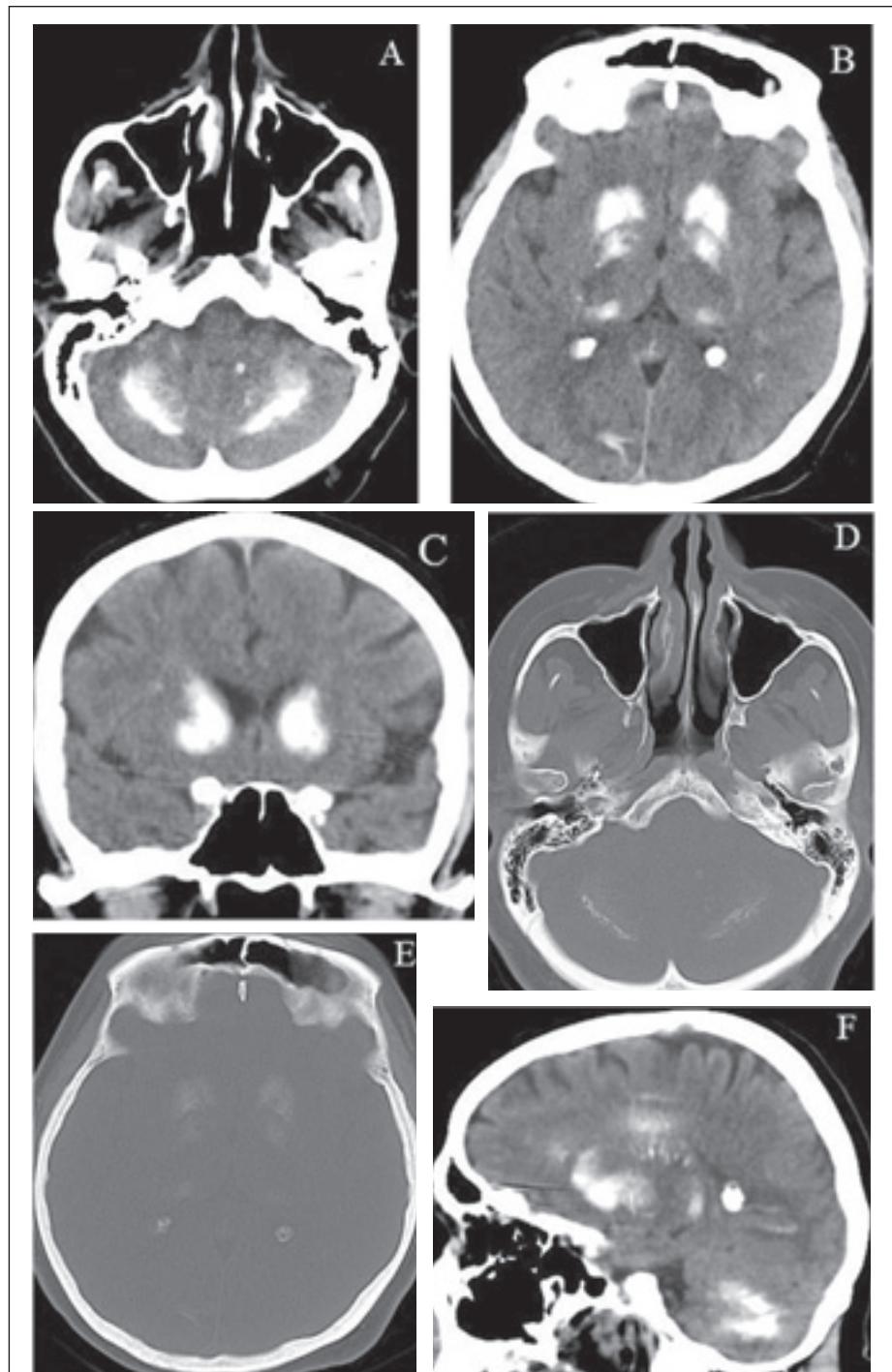


Fig. 1. Brain CT. (A, B) Axial views, (C) coronal view, (F) sagittal tissue view, (D, E) axial bone window. Bilateral symmetric hyperdense calcification areas.

Obr. 1. CT mozku. (A, B) Axialní pohledy, (C) koronární pohled, (F) sagitální pohled na tkáň, (D, E) axiální kostní okno. Bilaterální symetrické hyperdenzní kalcifikované oblasti.

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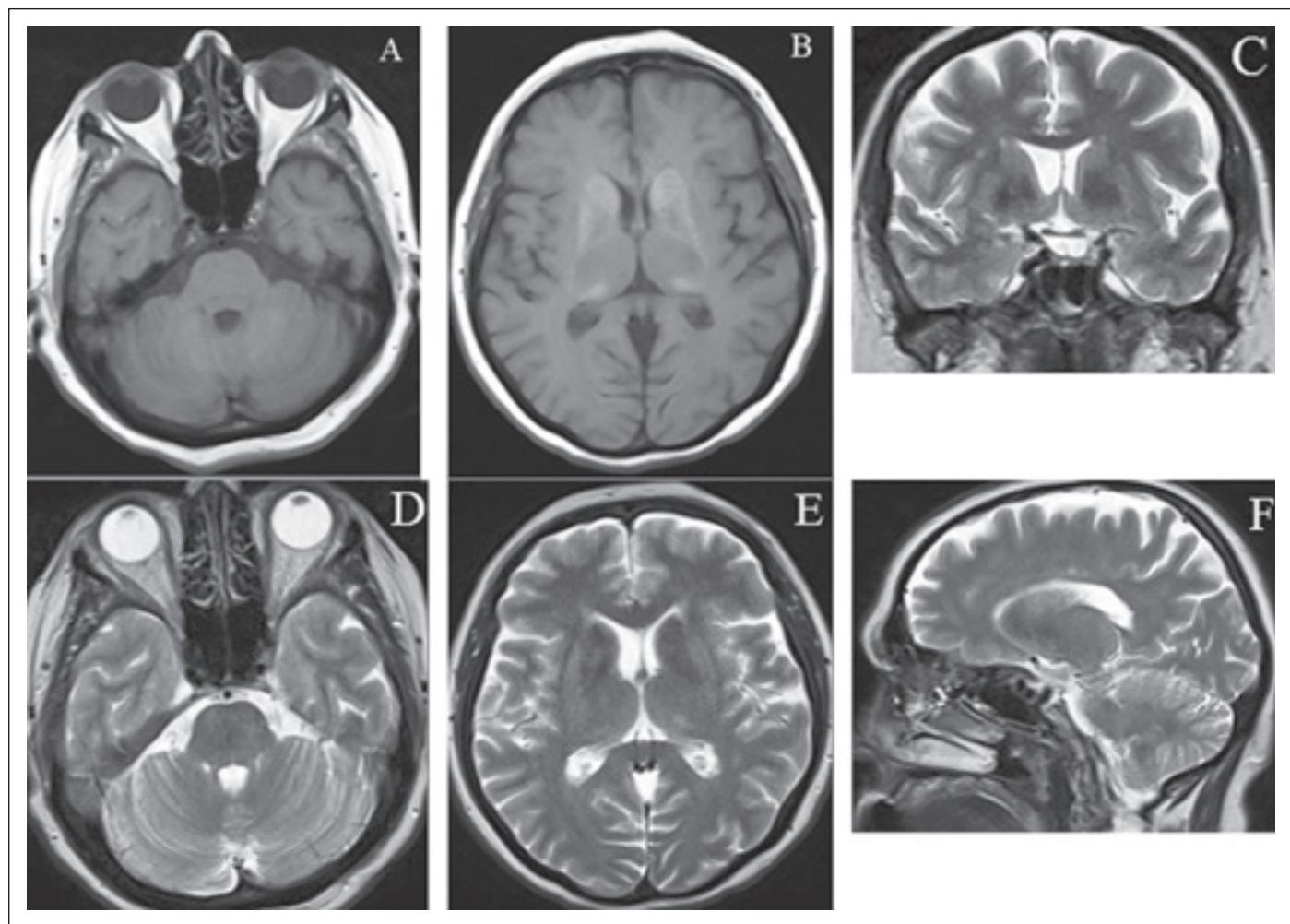


Fig. 2. Brain MRI. (A, B) Axial T1-WI images showing bilateral symmetric hyperintense areas, (C) coronal T2-WI, (D, E) axial T2-WI, (F) sagittal T2-WI showing bilateral symmetric hypointense areas.

Obr. 2. MRI mozku. (A, B) Axiální T1-WI snímky zobrazující bilaterální symetrické hyperintenzivní oblasti, (C) koronální T2-WI, (D, E) axiální T2-WI, (F) sagitální T2-WI zobrazující bilaterální symetrické hypointenzivní oblasti.

ric disturbances [3,4]. The increasing use of modern neuroimaging has resulted in incidental discovery of basal ganglia calcifications in up to 0.6% of brain CT scans [5].

We report the case of a 59-year-old female admitted to the emergency department after a motor vehicle accident. She had a medical history of right-sided congenital hip dislocation, long-standing arterial hypertension, diabetes mellitus, and a remote total thyroidectomy performed four decades earlier for multinodular goiter. Her medications included amlodipine, metformin, and levothyroxine. At presentation she had no neurological or psychiatric complaints. Physical examination revealed a thyroidectomy scar, and neurological examination was unremarkable, with no movement disorder or parkinsonian signs. The Mini-Mental States Examination yielded a score of 19, indicating

moderate cognitive impairment. Laboratory tests including complete blood count, biochemistry, and thyroid hormone levels were within normal limits, as were calcium and parathyroid hormone levels.

Cranial CT demonstrated bilateral symmetric calcifications in the basal ganglia, thalami, and cerebellum (Fig. 1). MRI confirmed iso- to hyperintense lesions on T1-weighted images and hypointense lesions on T2-weighted images in the caudate nuclei, pericallosal white matter, thalami, and cerebellar hemispheres (Fig. 2). No family history of neurological or psychiatric illness was present. Based on the neuroimaging, laboratory, and clinical findings, the diagnosis of Fahr's disease was established. The patient was followed up conservatively, and after one year no new symptoms developed.

Bilateral basal ganglia calcification was first reported by Bamberger in 1855 and clinically detailed by Fahr in 1930 [6]. Although autosomal dominant inheritance is most common, autosomal recessive inheritance and genetic loci on chromosome 14q have been implicated [7,8]. Pathologically, calcification involves the globus pallidus, putamen, caudate nucleus, internal capsule, thalamus, and cerebellar dentate nuclei [9]. Several mechanisms have been suggested, including local ischemia or inflammation leading to mineral deposition of calcium, iron, aluminum, copper, magnesium, and glycoproteins [9]. Another hypothesis proposes perivascular deposits due to degenerative vascular changes [9]. Similar radiological findings can occur in hypoparathyroidism, particularly after thyroidectomy, which is a frequent confounder in differential diagnosis [10]. In

our patient, normal calcium and parathyroid hormone levels excluded secondary metabolic causes, supporting a diagnosis of idiopathic Fahr's disease.

Although disease onset typically occurs in the fourth to sixth decades, calcifications may begin decades earlier. Clinical manifestations are highly variable and include parkinsonism, dementia, seizures, speech and gait disorders, and psychiatric symptoms ranging from depression to psychosis [9]. Nevertheless, some patients remain asymptomatic, and incidental discovery is not uncommon. In our case, aside from cognitive impairment, no major neurological or psychiatric manifestations were evident. This underscores the importance of considering Fahr's disease in incidental imaging findings even when clinical correlation is limited.

Imaging is critical for diagnosis. CT remains the most sensitive modality for detecting cerebral calcifications, while MRI provides anatomical localization but is less sensitive [9]. Incidental basal ganglia calcifications, especially in elderly patients, may reflect age-related vascular changes rather than Fahr's disease. Hypoparathyroidism – spontaneous or post-thyroidectomy – represents the most common secondary cause. Other differential diagnoses include infectious, metabolic, vascular, and congenital conditions such as tuberous sclerosis, toxoplasmosis, Cockayne syndrome, or prior cranial irradiation [9,10]. Careful clinical and laboratory correlation is therefore essential to distinguish idiopathic Fahr's disease from Fahr's syndrome.

There is currently no disease-modifying therapy available. Symptomatic treatments, such as antiparkinsonian agents or antiepileptics, may provide partial benefit. Trials of calcium channel blockers like nimodipine have been disappointing, possibly due to the mixed mineral composition of deposits [10]. Isolated reports suggest bisphosphonates such as disodium etidronate may reduce symptoms, but evidence remains anecdotal [10]. Given the lack of specific treatment, early recognition, differentiation from secondary causes, and long-term follow-up are of utmost importance.

This case highlights the importance of considering Fahr's disease in patients with widespread intracranial calcifications detected incidentally, even in the absence of overt neurological symptoms. Differentiation from secondary metabolic causes is essential, particularly in post-thyroidectomy patients. The incidental detection of Fahr's disease is likely to increase with the widespread use of CT, and awareness of its clinical and radiological features remains crucial for neurologists and neurosurgeons.

It should be emphasized that not all intracranial calcifications are benign age-related changes; in certain cases, they may reflect a progressive neurodegenerative condition requiring long-term follow-up. Therefore, clinicians should adopt a systematic approach that includes detailed medical history, laboratory assessment, and careful exclusion of metabolic causes to avoid misdiagnosis. Greater awareness may help prevent unnecessary interventions, ensure

appropriate counseling, and improve patient care in this rare but clinically significant disorder.

Conflict of interest

The authors declare they have no potential conflicts of interest concerning drugs, products, or services used in the study.

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