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Case Report

Fahr Syndrome: A Case Report of Asymptomatic Presentation

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Abstract

Fahr syndrome is often difficult to clinically suspect as it can remain asymptomatic or present with diverse manifestations that do not fit into a specific clinical profile. The predominant etiologies are dysparathyroidism, particularly hypoparathyroidism. Fahr disease can be genetic or sporadic. We report a case of a female patient who presented to our facility following a head injury, where a cerebral CT scan revealed intracerebral calcifications in the caudate, lenticular nuclei, and dentate nuclei bilaterally.

Keywords: Fahr Syndrome; Asymptomatic; Bilateral; Calcifications

Introduction

Bilateral intracerebral calcifications in the basal ganglia within Fahr syndrome or Fahr disease are rare entities. Fahr syndrome (FS), named after its description by Theodor Fahr in 1930, is radiologically defined by symmetrical, non-arteriosclerotic striato-pallidal and dentate calcifications [1].

Case Report

We describe the case of a 50-year-old female, with no significant medical history other than consanguineous marriage, who presented to our facility following a head injury with initial loss of consciousness. A cerebral CT scan was performed for lesion assessment, which revealed bilateral intracerebral calcifications in the caudate, lenticular nuclei, and dentate nuclei, with no post-traumatic lesions identified. Clinical examination was unremarkable, followed by further laboratory investigations: complete blood count showed no abnormalities, hypocalcemia was noted at 51 gm/L, and hypoparathyroidism with a parathyroid hormone level of 8 pg/ml.

Discussion

The idiopathic form, or Fahr disease, includes both sporadic and familial cases with autosomal dominant inheritance patterns. Recent studies have identified genetic mutations associated with primary familial brain calcification [2].

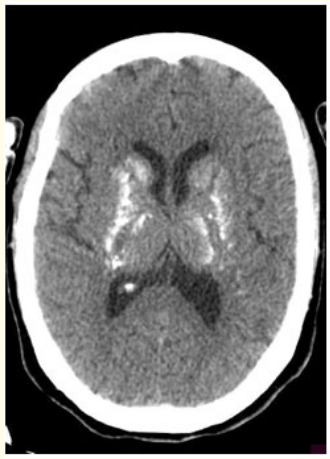

Figure 1: *Axial section of a brain scan showing calcifications of the caudate, lenticular and thalami nuclei.*

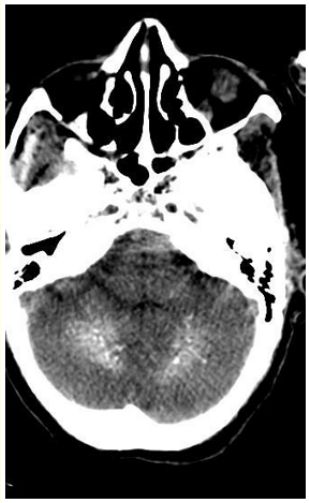

Figure 2: *Axial section of a brain scan showing calcifications of the dentate nuclei bilaterally.*

It primarily affects patients with dysparathyroidism, notably hypoparathyroidism [3]. The prevalence of Fahr syndrome is estimated at 0.5%. Pathophysiological mechanisms leading to intracerebral calcifications in FS are poorly understood, with hypotheses suggesting metabolic disturbances in oligoglia cells leading to mucopolysaccharide deposition and subsequent vascular, perivascular, and calcific plaque formation. These calcifications typically involve small vessels of the basal ganglia [4,5].

Clinical presentation varies widely, with many individuals remaining asymptomatic. Severe cases may manifest with parkinsonism, other movement disorders (e.g. chorea, dystonia), headaches, seizures and epilepsy, psychosis, depression, and progressive cognitive impairment [6,7].

CT is the preferred imaging modality for detecting intracerebral calcifications [8-10]. The most common locations include the striatum, thalamus, dentate nuclei, and centrum semiovale. In a meta-analysis of 19,080 cerebral CT scans, the overall incidence of bilateral and symmetrical calcifications in the basal ganglia was 6.6 per 1000 scans [11]. MRI, while less informative for FS, typically shows T1 and T2 hyperintensities, although T1 hypointensity may also occur, reflecting varying stages of evolution and differing chemical composition, including mucopolysaccharide content.

There is no specific treatment for FS apart from managing associated phosphocalcic disturbances, which may lead to partial improvement in neuropsychiatric symptoms [12].

Conclusion

Fahr syndrome is a rare entity diagnosed through clinical, laboratory, and radiological evaluations. Intracerebral calcifications should be considered in the presence of phosphocalcic metabolism disorders, particularly in cases with neurological or endocrine pathologies. Management focuses on correcting phosphocalcic imbalances.

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