

■ Case Report

Fahr's disease: a rare neurodegenerative disorder

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Abstract

Fahr's disease is a rare neurodegenerative disorder that is characterized by the bilateral symmetrical deposition of calcium in the basal ganglia, thalamus, dentate nuclei and centrum semiovale in the absence of hypoparathyroidism. It is often familial. Hereby, we are reporting a rare case of Fahr's disease who presented with repeated episodes of seizures and was diagnosed as Fahr's disease after performing computed tomographic (CT) scan.

Keywords: Fahr's disease, bilateral striopallidodentate calcinosis, familial, basal ganglia, calcification

Introduction

Fahr's disease is a rare degenerative neurological disorder characterized by the presence of abnormal calcium deposition and associated cell loss in the areas of brain that control movement, including basal ganglia and cerebral cortex.¹⁻⁴ The German pathologist Karl Theodor Fahr first described it in 1930.⁵ According to reports in medical literature, Fahr's disease is often familial. It is believed to have autosomal dominant inheritance but a few cases have been reported to have autosomal recessive inheritance and even some sporadic cases have been reported in literature.⁶ The association between the abnormal phenotypes and abnormal genes remain unclear despite the recent mapping to chromosome 14q of a susceptible locus for Fahr's disease.⁶

Case description

A 18 year-old boy with a history of repeated episodes of seizures and progressive deterioration of mental and motor functions presented to the department of radiodiagnosis for CT scan. He also complained of being "slow moving" for few years. There was no family history of seizure, tremor or other movement disorders. On examination, his verbal responses were

slow and he had impaired short-term memory. Axial plain CT scan of head was done which revealed extensive calcifications involving bilateral globus pallidus, putamen, caudate nucleus, internal capsule, thalami, dentate nucleus, cerebellum and subcortical white matter. Laboratory investigations were within normal limits. He had normal blood levels of parathyroid hormone, thyroid stimulating hormone, calcium, phosphate, iron studies and ceruloplasmin. The CT scan findings when correlated with typical clinical history and normal blood chemistry was suggestive of Fahr's Disease.

Discussion

Fahr's disease (also known as bilateral striopallidodentate calcinosis) is a rare neurodegenerative disease that is characterized by the bilaterally symmetric deposition of calcium (and other minerals) in the basal ganglia, thalamus, dentate nuclei, and centrum semiovale in the absence of hypoparathyroidism. It is not clear whether the calcification in Fahr's disease is a metastatic deposition, secondary to local disruption of blood brain barrier, or is due to disorder of neuronal calcium metabolism.³ Patients present with a slow onset of nonspecific symptoms such as headache, vertigo, movement disorders, syncope, and seizures. Other neurologic deficits include paresis, spasticity, gait disturbance, speech disorders, coma, dementia, Parkinsonism, chorea, tremors, dystonia, myoclonia,

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and orthostatic hypotension.⁷ Important alternatives in the radiologic differential diagnosis for Fahr's disease include hypoparathyroidism or pseudohypoparathyroidism (end-organ resistance to parathyroid hormone), which can be confirmed with measurements of serum calcium, phosphorus, and parathyroid hormone levels.^{8,9} Pseudohypoparathyroidism, in which there is no abnormality of calcium metabolism in asymptomatic patients, is another possible diagnosis in patients with widespread cerebral calcification. There is neither a cure for Fahr's disease, nor a standard course of treatment. The prognosis is variable and hard to predict¹⁰.

Conclusion

Fahr's disease should be included in the differential diagnosis of patients who presented with psychiatric symptoms associated with motor disorders when calcifications are found mainly in the basal ganglia. Making a clinical diagnosis of Fahr's disease relies on the combination of clinical features, brain imaging and exclusion of other causes of intracranial calcifications.

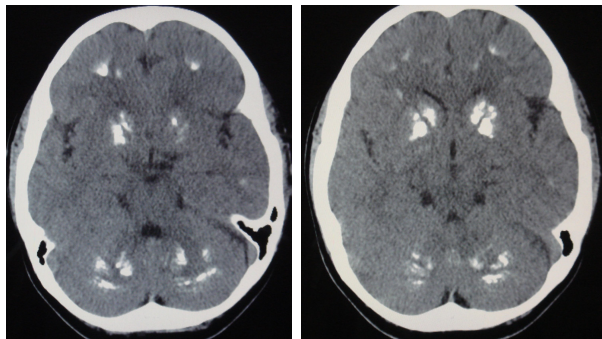


Figure 1 and 2. Axial plain CT scan of head shows extensive calcifications involving bilateral globus pallidus, putamen, caudate nucleus, internal capsule, dentate nucleus, cerebellum and subcortical white matter.

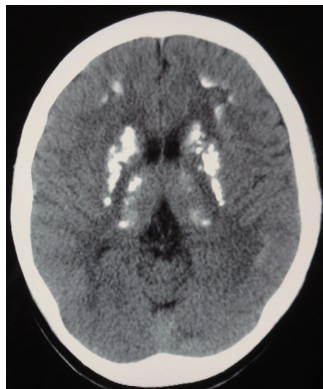


Figure 3. Axial plain CT scan of head shows extensive calcifications involving bilateral putamen, caudate nucleus, internal capsule, thalami and subcortical white matter.

References

1. Harrington M G, Macpherson P, McIntosh WB et al. The significance of the incidental finding of basal ganglia calcification on computed tomography. *J.Neurol. Neurosurg. Psychiat.* 44:-1168-1170, 1981
2. Smits MG, Gabreels FJ, Thijssen HOM et al. Progressive idiopathic strio-pallido-dentate calcinosis (Fahr's disease) with autosomal recessive inheritance: report of three siblings. *Europ. Neurol.* 22: 58-64, 1983
3. Malik R, Panday VK, Naik D. Fahr's disease. A rare neurodegenerative disorder. *Int J Radiol Image* 2004; 14:383-4
4. Hasan A, Bajpai G, Varshney N. Fahr's disease: case report. *J Ind Aca Clini Med* 2007; 8: 260-1
5. Fahr T. Idiopathische Verkalkung der Hirngefasse. *Zentralblatt für allgemeine Pathologie und pathologische Anatomie* 1930–1931;50:129–33
6. Geschwind DH, Loginov M, Stern JM. Identification of a locus on chromosome 14q for idiopathic basal ganglia calcification (Fahr disease). *Am. J. Hum. Genet.* 65: 764-772, 1999
7. Lauterbach EC, et al. Neuropsychiatric correlates and treatment of lenticulo-striatal diseases: a review of the literature and overview of research opportunities in Huntington's, Wilson's, and Fahr's diseases. A report of the ANPA Committee on Research. *American Neuropsychiatric Association. J Neuropsychiatry Clin Neurosci* 1998;10:249–66
8. Avrahami E, Cohn DF, Feibel M, Tadmor R. MRI demonstration and CT correlation of the brain in patients with idiopathic intracerebral calcification. *J Neurol* 1994; 241(6):381–384.
9. Smith AB, Smirniotopoulos JG, Rushing EJ, Goldstein SJ. Bilateral thalamic lesions. *AJR Am J Roentgenol* 2009; 192(2):W53–W62.
10. Brodaty H, Mitchell P, Luscombe G, et al. Familial idiopathic basal ganglia calcification (Fahr's disease) without neurological, cognitive and psychiatric symptoms is not linked to the IBGC1 locus on chromosome 14q. *Hum. Genet.* 110: 8-14, 2002