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Fahr Syndrome and Syncope: Case Report and Clinical Radiological Characteristics

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Abstract

Background: Fahr´s syndrome is a rare neurodegenerative entity, which consists of calcifications of the basal ganglia and cerebrospinal nuclei, which can be associated with neurological and neuropsychiatric symptoms. However, the difference between syndrome and Fahr's disease is highlighted.

Case Report: a 55-year-old man with underlying thyroid disease undergoing treatment, debuted with syncope with posterior cranial trauma, which was admitted to the emergency service, performing imaging studies with findings compatible with Fahr´s syndrome.

Conclusion: syncope as a cardiac symptom in a patient with Fahr syndrome, metabolic and structural abnormalities ruled out must be.

Keywords: Calcinosis; Syncope; Syndrome; Tomography; Fahr's syndrome.

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Introduction

Fahr syndrome/Fahr disease is a rare neurodegenerative disorder characterized by bilateral striatum-paleo-dentate calcinosis.[1,2] The first reports arise in 1850 by Delacourt who described calcifications in the basal ganglia in a man with weakness in the lower limbs. Later Bamberger described the calcium deposits in the cerebral vessels of a woman with epilepsy at the histopathological level in 1855. However, this pathology was recognized by Theodore Fahr, a German pathologist, in 1930 who documented the presence of cerebral calcification in an 81-year-old man with dementia. Fahr syndrome occurs with a prevalence of one per 100 000 habitants, with a male-female ratio of 2:1.[3]

Case Presentation

A 55-year-old male patient from a rural area, with a history of hypothyroidism in substitution, was admitted to the emergency department at the first level because of loss of consciousness associated with loss of postural tone with subsequent trauma to the cranial region, with spontaneous recovery after minutes, no relaxation of the sphincters. Upon re-examination, the patient reported syncope episodes lasting 1 year, for which an admission electrocardiogram was performed without alterations in the rhythm or conduction pathways. Later he was referred to a more complex center.

The patient was admitted, presented a sutured frontal wound, was assessed by the neurosurgery department with a Glasgow score of 14/15. Because of disorientation, a mini mental test was performed with a score of 18/30 points. Preserved cranial nerves, and their strength and sensitivity were normal. A CT scan (computed tomography of the skull) was performed with evidence of multiple calcifications in deep white matter, nuclei of the base and cerebellum hemispheres. No evidence of ischemic lesions or acute bleeding was seen (Figure 1). Given the imaging findings, a diagnosis compatible with Fahr's syndrome was made. Later, the internal medicine service followed up and monitored the patient. Given the patient's base condition, consent was obtained from family members for academic purposes.

Discussion

Fahr's disease and Fahr's syndrome, despite sharing similarities in signs and symptoms, vary with respect to their presentation, therapeutic approach, and etiological origin (Table 1). However, Fahr's disease has also received a series of names by which it has been described, such as: idiopathic basal ganglia calcification, bilateral dentate striated pale calcinosis and finally familial primary cerebral calcifications. All these definitions indicate a genetic disorder of neurodegenerative power associated with cerebral calcium deposition in the absence of secondary cause (Table 2).[3,4] However, up to 1.5% of the population may present with bilateral basal ganglion

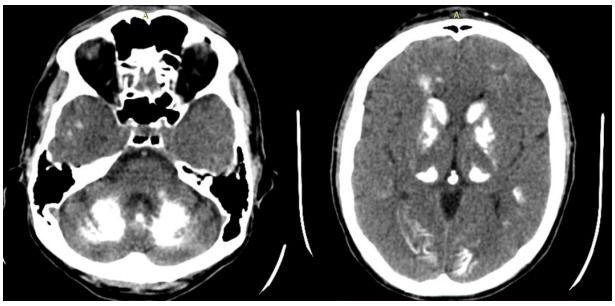

Figure 1. Computed Tomography: in Axial Slices, Evidence of Multiple Calcifications in Deep White Matter, Nuclei of the Base and Cerebellum Hemispheres.

Table 1. Main differential characteristics between Farh syndrome and Farh disease

	Fahr´s Syndrome	Fahr´s Disease
Age of onset	30 - 40 years	>40 years
Genetic disorder	None	Compromised gene: • SLC20A2 • PDGFRB • PDGFB • XPR1 • MYORG
Radiographic finding	Symmetrical and bilateral intracranial calcifications	
Secondary causes	Endocrinopathies Infections Autoimmune diseases Toxics Drugs	None
Associated syndrome	Cockayne tipo I y II Syndrome Coat´s Syndrome Mitochondrial diseases Aicardi-Goutières Syndrome Neuroferritinopathy Tuberous sclerosis	None
Treatment	According to underlying etiology + Symptomatic	Symptomatic

Modified and adapted from Ooi et al.[4]

calcifications.[5]

Fahr syndrome is characterized histologically by symmetric non-aromatous compounds embedded in a protein-polysaccharide complex that are located in the globe pallidus, striatum, dentate nucleus, and basal ganglia, as well as white matter and cerebellum. These deposits are made up of a series of elements, most of which is calcium, as well as zinc, iron, magnesium, copper, and phosphorus, taking into account that they adhere more to brain structures that are in greater contact with blood vessels.[6] Causing astrocytes and microglia to react and surround calcium deposits, thus organizing an inflammatory process.[7]

In neuroimaging, hyperdense lesions on brain tomography easily identify calcifications; these calcifications have a predilection for dentate nuclei and basal ganglia; however, they can be found in the thalamus, brainstem, semioval center, and subcortical white matter.[2] The gold standard with the best visualization of calcium deposits is still brain tomography, all due to multiple case reports where magnetic resonance imaging does not clearly show the presence of cerebral calcinosis.[8]

Conclusion

Fahr syndrome and Fahr disease are two similar conditions that share imaging characteristics, with computed This syndrome is accompanied by an insidious, slow onset that progressively increases between the fourth and sixth decade. However, the clinical presentation varies in a wide range of neurological or neuropsychiatric symptoms such

Table 2. Differences in cardiovascular findings and laboratory profile.

	Echocardiograma	Electrocardiogram	TSH	T4L	Ca+	Mg+	Phosphate
Broncel M. et al. (9)	hypokinesis of LV and systolic dysfunction	-Flattened T-waves in limb leads -Biphasic T-waves in precordial leads V3-V5 -Prolonged QTc interval (531 ms)	12,3uUI/ml	0,62ng/dl	3,6 mg/dl	None	7,3mg/dl
Pasca M. et al (10)	Negative for any dysfunction	- QTc interval (430 ms)	4uUI/ml	1,5 ng/dl	5,4mg/dl	1,78mg/dl	10,6mg/dl
Avellaneda L. et al (Our Study)	Negative for any dysfunction	-QTC interval (440 ms)	1.9mUI/ml	1,3 ng/dl	3,5 mg/dl	1,43mg/dl	None

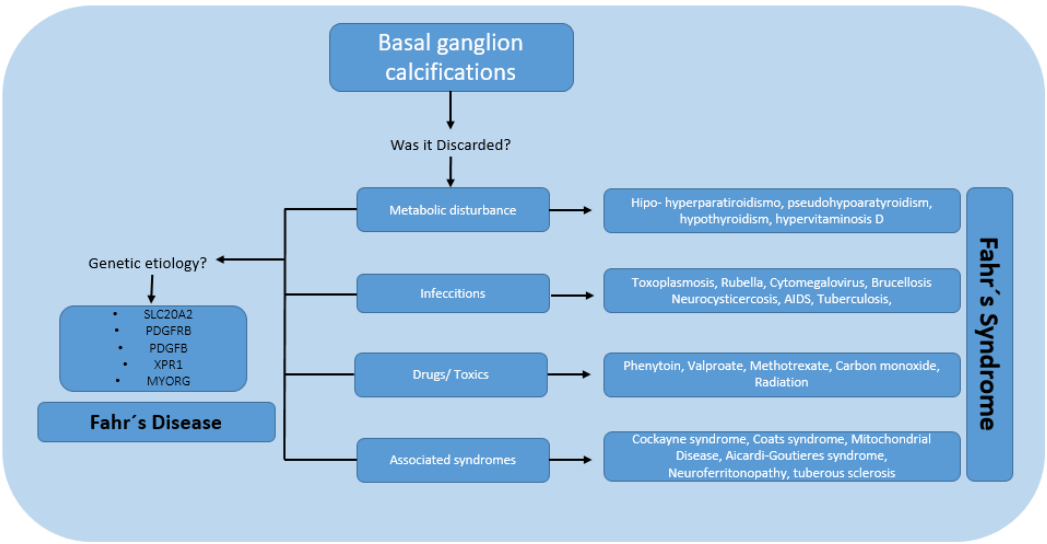

Figure 2. Algorithmic scheme in Fahr syndrome and Fahr disease.

as neurological (seizures, movement disorders, pyramidal signs, vertiginous symptoms, cerebellar symptoms, sensory disturbances, gait abnormalities) and neuropsychiatric (Cognitive alteration, psychotic symptoms, personality alterations, mood disorders, anxiety, and obsessive behavior) symptoms.[3,4,6] On the other hand, syncope as a cardiac manifestation is usually associated with uncompensated base metabolic pathologies, such as hypothyroidism or hypoparathyroidism. Despite this, structural or functional pathology must always be ruled out at the myocardial level (Table 2).[9,10] The management strategies for Fahr's syndrome and brain calcifications will depend on the patient's symptoms as well as the underlying etiology.[1-3] tomography being the study of choice to determine calcifications at the basal ganglia and cerebellar nuclei. However, they differ significantly with respect to management and etiological approach. The diagnostic process should be focused on determining the possible underlying causes of calcifications at the brain level and thus directing their respective treatment (Figure 2). It should be noted that syncope as a form of presentation is a rare way to debut, however, neurological or neuropsychiatric symptoms, as in this case, can be seen during the evaluation of these patients.

Conflict of Interest Disclosures

The authors declare that they have no conflict of interests.

Ethical Statement

Written informed consent was obtained from the patient for the publication of this report.

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