International Journal of Clinical and Medical Case Reports

ISSN 2690-1447

Case Study

Late-Onset Focal Chorea in Fahr's Disease Resulting From SLC20A2 Gene Mutation

Joseph Bruno Bidin Brooks[1], Fábio César Prosdócimi[1], Rodrigo André Oliveira[2], Guilherme Lopes da Silveira[2], Marcelo Abdul Nour Pechutti[3] and Amanda Vilela Gaiotto[3]*

[1]*Department of Structure and Function, Metropolitan University of Santos, Brazil*

[2]*Clínica MegaImagem-Santos, São Paulo, Brazil*

[3]*Department of Neurology, Irmandade Santa Casa Misericórdia de Santos, Brazil*

Introduction

Fahr's Disease (FD) is a rare autosomal dominant disease characterized by calcifications of the central nervous system. Clinical manifestation can occur in any age group, including late-onset disease in individuals over 40 years of age. Symptoms may be heterogeneous and commonly affect cognitive, psychiatric and motor domains, including movement disorders, such as, akinetic parkinsonism, ataxia, dystonia, and chorea. Other symptoms may be present, such as, seizures and chronic migraine. The diagnosis is obtained through detailed investigation of the clinical history, neuroimaging, laboratory and genetic evaluation [1-5].

Methods

The report of this case was submitted and approved by the ethics committee of the Metropolitan University of Santos.

Case Presentation

The present case refers to 50 year-old Caucasian male with a 2-year history of insidious and progressive cerebellar ataxia and choreic movement in the right upper limb. Personal and family medical background was unremarkable. Brain Computed Tomography (CT) scans showed symmetric calcification of the basal ganglia and thalami and asymmetric cerebellar involvement (Figure 1). Laboratory and cerebrospinal fluid tests showed results within the normal range. The genetic study showed mutation in the SLC20A2 gene and FD diagnosis was made. Symptomatic treatment was instituted with partial improvement of symptoms.

The mineralization and calcification process in FD can be triggered by several etiologies, including genetic mutations, as evidenced in the present study.

These case reports emphasize the importance of the clinical-radiological correlation in FD diagnosis, highlighting the relevance of the genetic study to elucidate the etiology of the disease and provide appropriate treatment.

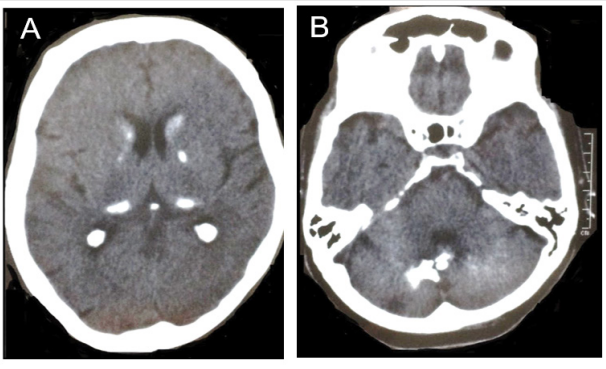

Figure 1: CT scan- Extensive and diffuse calcification of the basal ganglia (A) and cerebellum (B).

References

1. Carecchio M, Barzaghi C, Varrasi C, Cantello R, Garavaglia B. Adult-onset focal chorea in Fahr's disease resulting from SLC20A2 mutation: a novel phenotype. Mov Disord Clin Pract. 2014;2(1):79-80.

2. Hsu SC, Sears RL, Lemos RR, Quintáns B, Huang A, Spiteri E, et al. Mutations in SLC20A2 are a major cause of familial idiopathic basal ganglia calcification. Neurogenetics. 2013;14(1):11-22.

3. Baker M, Strongosky AJ, Sanchez-Contreras MY, Yang S, Ferguson W, Calne DB, et al. SLC20A2 and THAP1 deletion in familial basal ganglia calcification with dystonia. Neurogenetics. 2014;15(1):23-30.

4. Nicolas G, Pottier C, Charbonnier C, Guyant-Maréchal L, Le Ber I, Pariente J, et al. Phenotypic spectrum of probable and genetically-confirmed idiopathic basal ganglia calcification. Brain. 2013;136(pt 11):3395-407.

5. Zhu M, Zhu X, Wan H, Hong D. Familial IBGC caused by SLC20A2 mutation presenting as paroxysmal kinesigenic dyskinesia. Parkinsonism Relat Disord. 2014;20(3):353-4.

Citation: Brooks JBB, Prosdócimi FC, Oliveira RA, Silveira GLD, Pechutti MAN, Gaiotto AV. Late-Onset Focal Chorea in Fahr's Disease Resulting From SLC20A2 Gene Mutation. Int J Clin Med Case Rep. 2019; 1(1): 1003.

Publisher Name: Medtext Publications LLC

Manuscript compiled: Dec 19th, 2019

***Corresponding author:** Joseph Bruno Bidin Brooks, Metropolitan University of Santos (UNIMES), Avenida Gen, Francisco Glicério 8, Santos, São Paulo, 11045-002, Brazil, Tel/Fax: 551332283400, E-mail: joseph3b@gmail.com