ISSN: 2690- 909X

Case Report

Advances in Neurology and Neuroscience

CADASIL in the Differential Diagnosis of Atypical Parkinsonism. Case Report.

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Submitted: 10 Jan 2022; **Accepted:** 10 Feb 2022; **Published**: 23 Feb 2022

Citation: *Joseph Bruno Bidin Brooks. (2022). CADASIL in the Differential Diagnosis of Atypical Parkinsonism. Case Report. Adv Neur Neur Sci, 5(1): 09-10.*

Abstract

Autosomal dominant cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is an inherited small vessel disease caused by mutations in the NOTCH3 gene. The clinical spectrum includes migraine, transient ischemic attacks and/or recurrent strokes, and cognitive impairment. The presence of signs and symptoms of Parkinsonism are poorly described in the phenotype of this disease. This case report was approved by the Ethics Committee of Universidade Metropolitana de Santos.

Case Report: *The present case refers to a 59-year-old female patient presenting with recurrent transient ischemic attacks, chronic migraine, depression and bilateral akinetic-rigid parkinsonism. Brain MRI showed diffuse and confluent lesions. The genetic study showed a de novo heterozygous pathogenic variant in the NOTCH3 gene. Symptomatic treatment with multiprofessional rehabilitation was instituted with partial improvement of symptoms.*

Keywords: CADASIL, Atypical Parkinsonism, Case Report.

Introduction

Autosomal dominant cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (CADASIL) is an inherited small vessel disease caused by mutations in the NOTCH3 gene. The clinical spectrum includes migraine, transient ischemic attacks and/or recurrent strokes, and cognitive impairment. The presence of signs and symptoms of Parkinsonism are poorly described in the phenotype of this disease [1, 2]. We report a female patient with atypical Parkinsonism and mutation in the NOTCH3 gene responsible for CADASIL.

Methods

The report of this case was submitted and approved by the ethics committee of Universidade Metropolitana de Santos.

Case Presentation

The present case refers to a 59-year-old female patient, who presented a clinical picture of insidious and progressive onset 11 months before the evaluation and which was characterized by chronic migraine, depression and parkinsonism; Associated with the clinical picture described, the patient presented recurrent transient ischemic attacks (4 episodes), characterized by dysfunction in the language expression and lasting for 1 minute with complete recovery.

The neurological examination showed bilateral akinetic-rigid Parkinsonism, associated with postural instability - Modified Hewn Yahr Scale-3.0; United Parkinson's Disease Rating Scale (UPDRS)-52 points. Family history associated with Parkinsonism and dementia syndrome Figure 1.

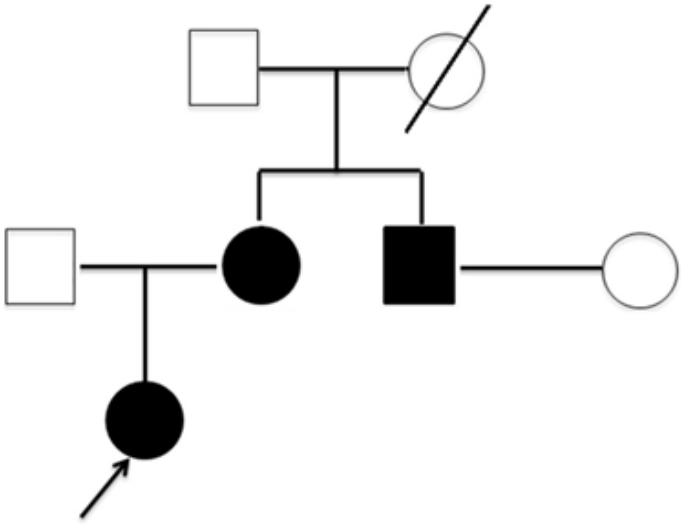

Figure 1: Family medical genogram. The index case has an indicator arrow. Symptomatic individuals for CADASIL are shown in black.

Complementary laboratory tests, as well as investigation for immune-mediated diseases, showed no changes. Brain magnetic resonance imaging showed diffuse and confluent lesions in the white matter, periventricular region, external capsule and basal ganglia Figure 2.

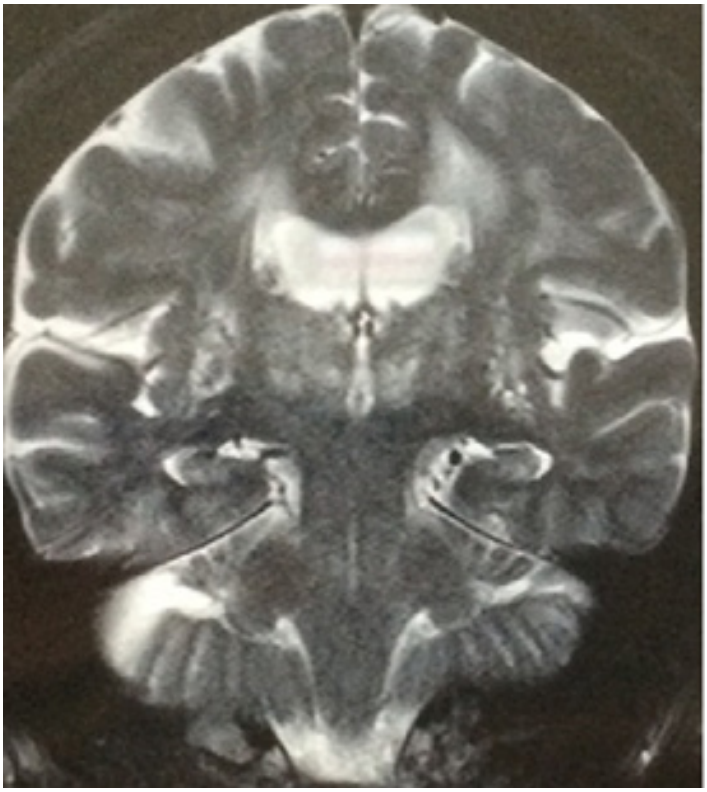

Figure 2: Brain- T2-MRI- Leukoencephalopathy characterized by diffuse and confluent hyperintense lesions in the white matter, periventricular region, external capsule and basal nuclei.

The variant Chr19:15298704 G>A was identified in heterozygosity, in the NOTCH3 gene, (OMIM #600276), which promotes the substitution of the amino acid arginine at codon 532 by cysteine (p.Arg532Cys).

The finding of the pathogenic variant in the NOTCH3 gene was postulated to be associated with this patient's clinical condition.

Multiprofessional treatment was performed with physiotherapy, speech therapy, occupational therapy and psychology. Drug treatment was instituted with levodopa and with the dose of 800 mg daily with a suboptimal response.

Clinical and rehabilitation treatment resulted in partial improvement of neurological symptoms.

Conclusion

Autosomal dominant cerebral arteriopathy with subcortical infarcts and leukoencephalopathy is a challenge due to the heterogeneity of clinical presentation. With the advent and development of neurogenetics, many etiologies were then better understood. The study and inclusion of the NOTCH3 gene should be encouraged in patients with recurrent transient ischemic attacks, migraine, dementia and atypical Parkinsonism.

Author Disclosures

Joseph Bruno Bidin Brooks declares that he has no conflict of interest.

References

1. Valenti, R., Bianchi, S., Pescini, F., D'Eramo, C., Inzitari, D., Dotti, M. T., & Pantoni, L. (2011). First report of a pathogenic mutation on exon 24 of the NOTCH3 gene in a CADASIL family. Journal of neurology, 258(9), 1632-1636.
2. Wegner, F., Strecker, K., Schwarz, J., Wagner, A., Heinritz, W., Sommerer, F., & Sabri, O. (2007). Vascular Parkinsonism in a CADASIL case with an intact nigrostriatal dopaminergic system. Journal of neurology, 254(12), 1743-1745.