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Manuscript Type: CASE REPORT

A novel frameshift mutation in the SACS gene causing spastic ataxia of charlevoix-saguenay in a consanguineous family from the Arabian Peninsula: A case report and review of literature

Abstract

BACKGROUND

Familial cases of autosomal recessive spastic ataxia of charlevoix-saguenay have not been reported in the Arabian Peninsula, although the consanguineous marriage rate is very high. We report the first family from the Arabian Peninsula harboring a novel frameshift mutation in the SACS gene.

CASE SUMMARY

Match Overview

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Cited by: 135

Author: Suman Jayadev, Thomas D. Bird

Publish Year: 2013

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This chapter presents genetic testing for hereditary **ataxia** and hereditary **spastic** paraplegia. It focuses on molecular diagnostic testing for the degenerative ataxias, which are often clinically indistinguishable from each other and for which genetic testing provides the only means to a specific diagnosis.

Author: Martha A. Nance

Publish Year: 2007



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Autosomal recessive spastic ataxia of Charlevoix ... 翻译此页

The finding of a novel homozygous mutation - c.3066del(p.Asn1025Metfs*10) - in the SACS gene, resulting in a frameshift and a premature stop codon, allowed the genetic confirmation of the clinical diagnosis. Key words. ataxia of Charlevoix-Saguenay, SACS, early-onset ataxia, spastic paraparesis, polyneuropathy. Introduction

<https://www.oatext.com/Autosomal-recessive-spastic...>

Novel SACS mutations in autosomal recessive spastic ... 翻译此页

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Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is a distinct form of hereditary early-onset spastic ataxia. In 2000, the causative gene, SACS, encoding the protein saccin, was ...

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SPASTIC ATAXIA, CHARLEVOIX-SAGUENAY TYPE; ... 翻译此页

A number sign (#) is used with this entry because spastic ataxia of the Charlevoix-Saguenay type (SACS, or ARSACS) is caused by homozygous or compound heterozygous mutation in the gene encoding the saccin protein (SACS; 604490) on chromosome 13q12 For a discussion of genetic heterogeneity of spastic ataxia, see SPAX1 (). Description

<https://www.omim.org/entry/270550>





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Molecular diagnosis of autosomal recessive cerebellar ...

<https://www.wjgnet.com/2218-6212/full/v3/i4/115.htm>

Another **novel** homozygous **mutation** in **SACS** (c.2439-2440delAT [p.Val815Glyfs*4]) has been identified in a Tunisian **family** with two affected individuals, by homozygosity mapping and WES. A more recent study on a 4-year-old girl revealed an additional **novel frameshift** homozygous **mutation** in the same **gene** ...

Cited by: 2

Author: Christina Votsi, Kyproula Christodoulou

Publish Year: 2013

Chapter 18 Genetic Testing for Hereditary Ataxia and ...

<https://www.sciencedirect.com/science/article/pii/S1877184X09700929>

This chapter presents genetic testing for hereditary **ataxia** and hereditary **spastic** paraplegia. It focuses on molecular diagnostic testing for the degenerative ataxias, which are often clinically indistinguishable from each other and for which genetic testing provides the only means to a specific diagnosis.

Author: Martha A. Nance

Publish Year: 2007

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