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Relating Variation to Medicine

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Overview

RCV000000001.1 Last Updated: Oct 29, 2010

A patient with Farber lipogranulomatosis was homozygous for a missense variant ASN320ASP (N320D) in ASAH1 resulting from a 958A>G single nucleotide variant. Loss of function was verified in cultured cells. (PubMed 11241842)

Number	Phenotype	Variant	Relationship	PubMed show details
1	Farber lipogranulomatosis	N320D	likely pathogenic	11241842

Try also

- Find this variant in PubMed
- See all medically related variants for ASAH1
- See all variants associated with Farber lipogranulomatosis
- Find other resources with information about Farber lipogranulomatosis

Variation

Variation type: single nucleotide change
Molecular effect: missense

Names:

Type	Identifier	Name/expression	Go there
HGVS expression	RefSeqGene	NG_008985.1:g.30575A>G	☑
HGVS expression	RefSeq mRNA	NM_177924.3:c.958A>G	☑
OMIM	613468.0004	ASAH1 ASN320AS	☑

Number	Details	
1	Ethnicity: Turkish female	consanguinity Farber disease subtype 6 (11241842)

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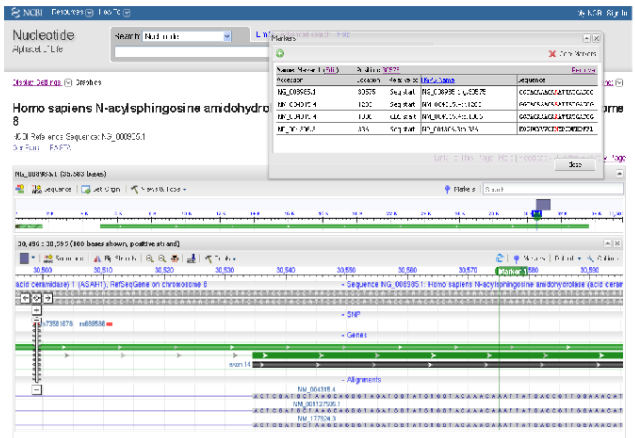
Phenotype

Name: Farber Lipogranulomatosis, UMLS C0268255
Alternates: FARBER LIPOGRANULOMATOSIS, OMIM 228000
Ceramidase deficiency

Related citations:
GeneReviews - none
Orphanet - none
Reviews in PubMed Park JH, Schuchman EH, Biochim Biophys Acta. 2006 Dec;1758(12):2133 8

Genomic context



Submissions

SCV000000001.1 Submitted: October 29, 2010
Asserted relationship between Farber lipogranulomatosis and NG_008985.1:g.30575A>G was extracted from PubMed 11211842 and mapped to the RefSeqGene by NCBI staff.

Genetic Testing Registry

Report from Genetic Testing Registry summarizing tests for this variation