

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.1067C>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 5	M1	Uncertain significance

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M ₁ orléans		Unknown	p.Ala356Asp
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
			M
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
0.71		11860	

Comments

associated with a M1 allele.

Occurrence

Ethnic background without frequency range : French Caucasian

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)
Occurrence comments			
Overall comments			
Occurrence comments			
This variant was identified at heterozygous status in a 62-year old man presenting with liver steatosis associated with hyperferritinemia.			
Last Update			
First publication : 07-23-2020 12:38 Last update : 08-08-2020 07:03 by Pr Curateur test			