

### Mutation sequence analysis

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HGVS nomenclature (NM\_000295.4)

Nomenclature including the signal peptide

c.879C>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M1	Uncertain significance

### Comments

rs141095970

### AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M <sub>1</sub> steenwerck		Unknown	p.His293Gln
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
			M
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
<b>Heterozygous</b>	<b>Homozygous</b>	<b>Heterozygous</b>	<b>Homozygous</b>
1.57		20858	

### Comments

associated with a M1 allele.

### Occurrence

Ethnic background without frequency range :

### Ethnic background and frequency

Frequency range		Group tested	
from (%)	To (%)	Size	Description (who was tested)
0.01	0.18		
<b>Occurrence comments</b>			
from gnomAD (2.1)			
<b>Overall comments</b>			
<b>Occurrence comments</b>			
This variant was identified at an heterozygous status with a M1 allele in a 32-year old woman presenting a severe hepatic cytolysis and cholestasis in a systemic lupus background.			
<b>Last Update</b>			
First publication : 07-16-2020 16:31 Last update : 08-12-2020 18:14 by Pr Curateur test			