

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.211A>C

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1	Likely benign

Comments

rs11575873

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
S _{Roubaix}		Unknown	p.Ser71Arg
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
beta-sheet 6	W		S
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
1.29		18314	

Comments

Associated with a M2 allele anti-elastasic activity within the reference values (17500-31500 UI/L)

Occurrence

Ethnic background without frequency range : North African

Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
		50/282542	gnomAD	
Occurrence comments				
Frequency in overall population				
Overall comments				
Occurrence comments				
post HCV cirrhosis				
References				
Medline ID	Authors		Title	
30223862	Renoux C, Odou MF, Tosato G, Teoli J, Abbou N, Lombard C, Zerimech F, Porchet N, Chapuis Cellier C, Balduyck M, Joly P		Description of 22 new alpha-1 antitrypsin genetic variants.	
Journal	Year	Volume	Num	Pp
Orphanet journal of rare diseases	2018	13	1	161
Last Update				
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