

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.1200A>C

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 5	M3	Uncertain significance

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M ₃		Precipitating	p.Glu400Asp
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
0.67		9276	

Comments

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)		
11.07	42.52				
Occurrence comments					
From gnomAD (2.1)					
Overall comments					
Occurrence comments					
This variant was identified at a heterozygous status with a S allele in a 48-year old man presenting with severe cirrhosis associated with a NASH syndrome and waiting for a liver transplantation.					
References					
Medline ID	Authors			Title	
32427833	Bashir A,Hazari Y,Pal D,Maity D,Bashir S,Singh LR,Shah NN,Fazili KM			Aggregation of M3 (E376D) variant of alpha1- antitrypsin.	
Journal		Year	Volume	Num	Pp
Scientific reports		2020	10	1	8290
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