

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.227_229delTCT

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

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M _{palermo}		Deficient Precipitating	p.F76del
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
	0.17		4428

Comments

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)		
	0.04				
Occurrence comments					
from gnomAD (2.1)					
Overall comments					
Occurrence comments					
This variant was identified at a homozygous status in a 49-year old woman presenting with COPD associated with emphysema.					
References					
Medline ID	Authors		Title		
7977369	Faber JP,Poller W,Weidinger S,Kirchgesser M,Schwaab R,Bidlingmaier F,Olek K		Identification and DNA sequence analysis of 15 new alpha 1-antitrypsin variants, including two PI*Q0 alleles and one deficient PI*M allele.		
Journal		Year	Volume	Num	Pp
American journal of human genetics		1994	55	6	1113-21
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