

Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.682G>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M2	Benign

Comments

rs199422208

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
X		Neutral	p.Glu228Lys
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
	X		X
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
1.3			

Comments

AAT concentration reported on a single case-report in a man in the Lyon area.

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
Occurrence comments				
Overall comments				
Occurrence comments				
References				
Medline ID	Authors		Title	
11524735	Lodewyckx L,Vandevyver C,Vandervorst C, Van Steenberg W,Raus J,Michiels L		Mutation detection in the alpha-1 antitrypsin gene (PI) using denaturing gradient gel electrophoresis.	
Journal	Year	Volume	Num	Pp
Human mutation	2001	18	3	243-50
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