

Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.592A>G

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

Comments

rs22008137

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
F _{lyon}		Neutral	p.Lys198Glu
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
	F		F
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
1.75			

Comments

No supplementary band for this AAT variant contrary to the F-variant.

Occurrence

Ethnic background without frequency range : French

Ethnic background and frequency

Frequency range		Group tested			
from (%)	To (%)	Size	Description (who was tested)		
Occurrence comments					
Discovered in a man living in Lyon.					
Overall comments					
Occurrence comments					
This variant seems to be neutral but more observations would be needed for a definitive conclusion.					
References					
Medline ID	Authors		Title		
22008137	Joly P, Francina A, Lacan P, Heraut J, Chapuis-Cellier C		[Place of genotyping in addition to the phenotype and the assay of serum ?-1 antitrypsin].		
Journal		Year	Volume	Num	Pp
Annales de biologie clinique			69	5	571-6
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