

### Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM\_000295.4)

Nomenclature including the signal peptide

c.77A>C

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

### Comments

rs199422212

### AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
V <sub>munich</sub>		Neutral	p.Asp26Ala
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
	V		V
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous

### Comments

Unique IEF banding pattern in which the 7 and 8 alpha 1AT protein bands focus with the normal M-type 7 and 8 bands, despite the fact that the major fraction of the V<sub>munich</sub> protein focuses in the "V" region of the IEF gel

### Occurrence

Ethnic background without frequency range : German

### Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
<b>Occurrence comments</b>				
<b>Overall comments</b>				
<b>Occurrence comments</b>				
<b>References</b>				
Medline ID	Authors	Title		
2316526	Holmes MD,Brantly ML,Curiel DT,Weidinger S,Crystal RG	Characterization of the normal alpha 1-antitrypsin allele Vmunch: a variant associated with a unique protein isoelectric focusing pattern.		
Journal	Year	Volume	Num	Pp
American journal of human genetics	1990	46	4	810-6
<b>Last Update</b>				
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