Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM_000295.4)

Nomenclarure including the signal peptide

c.739C>T

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M1 Val	Pathogenic

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
F		Dysfunctional	p.Arg247Cys

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**
1.02-2.23		13900-18900	

Comments

In the context that the normal alpha 1AT molecule has only one cysteine residue, a mutation resulting in the addition of a second cysteine may influence the three-dimensional form of the protein and/or permit interaction with other plasma proteins with free-SH groups and may be responsible for the observation that the major F alpha 1AT bands often migrate as doublets in IEF gels

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
2035534	Okayama H,Brantly M,Holmes M,Crystal RG	Characterization of the molecular basis of the alpha 1-antitrypsin F allele.

Journal	Year	Volume	Num	Pp
American journal of human genetics	1991	48	6	1154-8

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