Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM_000295.4)

Nomenclarure including the signal peptide

c.1061C>T

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 4	M1 Val	Benign

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
S_{munich}		Neutral	p.Ser354Phe

3D position of aa affected	Mobility on polyacrylamide gel	Mobility on agarose gel
	S	S Slow

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
1.60		16000-20600	

Comments

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
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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