

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.953delC

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 4	M1	Pathogenic

Comments

Genetic background : M1 or M2

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Q ₀ anzin		Deficient	p.Thr318Metfs*6
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
0.75		12524	

Comments

No band corresponding to the variant detectable in IEF on agarose gel.

Occurrence

Ethnic background without frequency range : European

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)
Occurrence comments			
Not reported in frequency databases			
Overall comments			
Occurrence comments			
Mutation identified in heterozygous status (with a PI*M allele) in a 62 year-old woman presenting with COPD associated with severe pulmonary emphysema.			
Last Update			
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