

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.288_291delTCAC

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 2	Other	Pathogenic

Comments

Genetic background: Z

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Q ₀ lille		Deficient	p.(His97Metfs*7)
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
1.40		18259	

Comments

Variant identified at heterozygous status M1Q0lille in inflammatory condition with a M IEF profile.

Occurrence

Ethnic background without frequency range : North African

Ethnic background and frequency

Frequency range	Group tested
-----------------	--------------

from (%)	To (%)	Size	Description (who was tested)		
Occurrence comments					
not referred in gnomAD (2.1)					
Overall comments					
Occurrence comments					
Variant identified at heterozygous status M1Q0lille in a 33-year old man presenting with recurrent pneumothorax.					
References					
Medline ID	Authors		Title		
30223862	Renoux C, Odou MF, Tosato G, Teoli J, Abbou N, Lombard C, Zerimech F, Porchet N, Chapuis Cellier C, Balduyck M, Joly P		Description of 22 new alpha-1 antitrypsin genetic variants.		
Journal		Year	Volume	Num	Pp
Orphanet journal of rare diseases		2018	13	1	161
Last Update					
First publication : 06-09-2020 11:22 Last update : 06-09-2020 11:35 by Pr test2 Compte					