

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.188G>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1	Uncertain significance

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M _{rouen}		Unknown	p.Arg63His
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
			M
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous

Comments

Biological data aren't available

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)		
0.00	0.00				
Occurrence comments					
from gnomAD (2.1)					
Overall comments					
Occurrence comments					
This variant was identified at a heterozygous status in a 39-year old man presenting with alpha1 antitrypsin deficiency.					
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					
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