

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.116A>T

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1	Likely benign

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M ₁ bruxelles			p.His39Leu
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
1.31		20731	

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				
This variant was identified at a heterozygous status in a 36-year old man presenting with emphysema and cholestatsis.				
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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