

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.1130dupT

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 5	M3	Pathogenic

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Q ₀ ourém		Deficient	p.Leu377Phefs*24
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
0.66		11879	

Comments

No band detectable on agarose gel. Mutation described as Q₀mattawa which occurs on a PI*M1 background.

Occurrence

Ethnic background without frequency range : Portuguese

Ethnic background and frequency

Frequency range	Group tested
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from (%)	To (%)	Size	Description (who was tested)		
Occurrence comments					
data from gnomAD (2.1)					
Overall comments					
Occurrence comments					
Variant identified at homozygous status in a 33 year-old man presenting with dyspnea on exertion, associated with a moderate airflow obstruction, incompletely reversible. The thoracic CT-scan showed emphysema.					
References n°1					
Medline ID	Authors		Title		
21457231	Vaz Rodrigues L,Costa F,Marques P,Mendonça C,Rocha J,Seixas S		Severe α -1 antitrypsin deficiency caused by Q0(Ourém) allele: clinical features, haplotype characterization and history.		
Journal		Year	Volume	Num	Pp
Clinical genetics		2012	81	5	462-9
References n°2					
Medline ID	Authors		Title		
26604020	Perrin J,Aimone-Gastin I,Balduyck M,Mercy M,Filhine-Trésarrieu P,Odou MF,Chaouat A,Chabot F		[Alpha-1 antitrypsin deficiency caused by Null mutation].		
Journal		Year	Volume	Num	Pp
Revue des maladies respiratoires		2016	33	7	612-7
Last Update					
First publication : 05-15-2020 11:29 Last update : 05-19-2020 12:37 by Pr test2 Compte					