

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.221T>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M3	Likely pathogenic

Comments

rs1275309068. This variant was initially described (but not named) by Carpagnano et al in 2017.

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
T _{ijarafe}		Deficient Precipitating	p.Ile74Asn
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
			T
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
0.78		11455	

Comments

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
Occurrence comments				
from gnomAD (2.1)				
Overall comments				
Occurrence comments				
This variant was identified at a heterozygous status in a 48-year old man presenting with pulmonary obstruction and emphysema.				
References n°1				
Medline ID	Authors			Title
29232161	Matamala N,Lara B,Gomez-Mariano G,Martínez S,Retana D,Fernandez T,Silvestre RA,Belmonte I,Rodriguez-Frias F,Vilar M,Sáez R,Iturbe I,Castillo S,Molina-Molina M,Texido A,Tirado-Conde G,Lopez-Campos JL,Posada M,Blanco I,Janciauskiene S,Martinez-Delgado B			Characterization of Novel Missense Variants of SERPINA1 Gene Causing Alpha-1 Antitrypsin Deficiency.
Journal	Year	Volume	Num	Pp
American journal of respiratory cell and molecular biology	2018	58	6	706-716
References n°2				
Medline ID	Authors			Title
28668972	Carpagnano GE,Santacroce R,Palmiotti GA,Leccese A,Giuffreda E,Margaglione M,Foschino Barbaro MP,Aliberti S,Lacedonia D			A New SERPINA-1 Missense Mutation Associated with Alpha-1 Antitrypsin Deficiency and Bronchiectasis.
Journal	Year	Volume	Num	Pp
Lung	2017	195	5	679-682
Last Update				
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