AVB-CG
CENTRE DE RÉFÉRENCE
ATRÉSIE DES VOIES BILIAIRES
ET CHOLESTASES GÉNÉTIQUES

Mutation sequence analysis			
Contributed by : CHU Lille			
HGVS nomenclature (NM_000295.4)			
Nomenclarure including the signal peptide			
c.1159G>A			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 5	M1	Uncertain significance
Comments			
rs121912712			
AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$X_{christchurch}$		Unknown	p.Glu387Lys

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**
1.49		24769	
Comments			
Associated with a M1 allele.			
Occurrence			
Ethnic background without frequency range : Asian			
Ethnic background and frequency			

Frequency range	**Group tested**

from (%)	To (%)	Size	Description (who was tested)
0.02	0.28		

Occurrence comments

From gnomAD (2.1)

Overall comments

Occurrence comments

This variant was identified at a heterozygous status in a 57-year old man presenting with severe pulmonary sarcoïdosis complicated with an episode of spontaneous pneumothorax. This variant was also identified at an heterozygous status MXchristchurch in a 29 year-old woman presenting with interstitial lung disease in the context of systemic lupus erythematosus and Gougerot Sjogren syndrome.

References

Medline ID	Authors	Title
3527273	Brennan SO,Carrell RW	alpha 1-Antitrypsin Christchurch, 363 Glu----Lys: mutation at the P'5 position does not affect inhibitory activity.

Journal	Year	Volume	Num	Pp
Biochimica et biophysica acta	1986	873	1	13-9

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