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.721A>T			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M1	Pathogenic
Comments			
rs199422211			
AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0bellingham}$		Deficient	p.Lys241*

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**
0.88		11857	
Comments			
associated with a M3 allele.			
Occurrence			
Ethnic background without frequency range :			
Ethnic background and frequency			

Frequency range	**Group tested**

from (%)	To (%)	Size	Description (who was tested)
	0.04		

Occurrence comments

from gnomAD (2.1)

Overall comments

Occurrence comments

This variant was identified at an heterozygous status with a M3 allele in a 6-year old presenting an immune deficiency.

References

Medline ID	Authors	Title
3257351	Satoh K,Nukiwa T,Brantly M,Garver RI Jr,Hofker M,Courtney M,Crystal RG	Emphysema associated with complete absence of alpha 1- antitrypsin in serum and the homozygous inheritance [corrected] of a stop codon in an alpha 1-antitrypsin-coding exon.

Journal	Year	Volume	Num	Pp
American journal of human genetics	1988	42	1	77-83

Last Update

First publication : 07-16-2020 16:45 Last update : 08-14-2020 17:48 by Pr Curateur test