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.922G>T			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 4	M1	Uncertain significance
Comments			
rs141620200			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M_{1Lyon}		Unknown	p.Ala308Ser

3D position of aa affected	Mobility on polyacrylamide gel	Mobility on agarose gel
		M

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
0.82		11099	
Comments			
associated with a Z allele.			
Occurrence			
Ethnic background without frequency range :			
Ethnic background and frequency			

Frequency range		Group tested	
from (%)	To (%)	Size	Description (who was tested)
0.01	0.39		

Occurrence comments

from gnomAD.

Overall comments

Occurrence comments

This variant was identified at a heterozygous status with a Z allele in a 43-year old woman (familial deficiency). This variant was identified at a heterozygous status M1LyonZ in a 10-year old girl presenting with cystic fibrosis requiring liver transplant. A familial screening was realised : her father was diagnosed M1lyonM2 with an AAT serum concentration of 1.14 g/L et her brother was diagnosed M1lyonS with an AAT serum concentration of 1.15 g/L(picture 2).

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

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