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.244G>A			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1	Uncertain significance
Comments			
rs113817720			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$M_{1Strasbourg}$		Unknown	p.Ala82Thr

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		12203	

Comments
associated with a S allele.
Occurrence
Ethnic background without frequency range :
Ethnic background and frequency

Frequency range		Group tested	
from (%)	To (%)	Size	Description (who was tested)
	0.16		

Occurrence comments
from gnomAD.

Overall comments
Occurrence comments
This variant was identified at an heterozygous status with a S allele in a 48-year old woman (familial deficiency).
Last Update
First publication : 07-16-2020 15:40 Last update : 08-08-2020 06:29 by Pr Curateur test