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

Mutation sequence analysis			
Contributed by : CHU Lyon			
HGVS nomenclature (NM_000295.4)			
Nomenclarure including the signal peptide			
c.185A>G			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1 Ala	Likely pathogenic
Comments			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
O_{feyzin}		Deficient	p.Tyr62Cys
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**
0.59			
Comments			

Occurrence			
Ethnic background without frequency range : Algerian			
Ethnic background and frequency			
Frequency range		**Group tested**	
from (%)	To (%)	Size	Description (who was tested)

Occurrence comments			
First description in a boy with neo-natal icterus and originating from Algeria.			
Overall comments			
Occurrence comments			
This new AAT variant is also very likely pathogenic considering in silico analyses: - very conserved amino-acid in evolution (phyloP = 4.81) - important Grantham distance between Tyr and Cys: 194 [0-215] - SIFT: deletorious score (score: 0.01; median: 2.73) - Mutation Taster: disease causing (p-value = 0.999)			
Last Update			
First publication : 12-24-2019 11:14 Last update : 02-02-2020 20:28 by Pr Curateur test			