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.1065+1G>A			
Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Intron 4	M1	Pathogenic
Comments			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0amiens}$		Deficient	p.?

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**
1.18		17419	
Comments			
AAT level probably determined in inflammatory condition			
Occurrence			
Ethnic background without frequency range : European			
Ethnic background and frequency			
Frequency range	**Group tested**		

from (%)	To (%)	Size	Description (who was tested)
0.00	0.00		

Occurrence comments

frequency from gnomAD (2.1) rs781591420

Overall comments

Occurrence comments

Fortuitous identification in heterozygous status (with a PI*M1 allele) in a 81 year-old woman with abnormal serum protein electrophoretic pattern. This mutation was also identified in heterozygous status (with a PI*M3 allele) in a 29 year-old woman presenting with a chronic neutropenia (AAT=0.67 g/L and anti-elastolytic activity =9750 UI/L)

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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