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

Mutation sequence analysis			
Contributed by : CHU de Lille			
HGVS nomenclature (NM_000295.4)			
Nomenclarure including the signal peptide			
c.1158dup			
Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 5	M1 Val	Pathogenic
Comments			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0clayton}$		Deficient	p.(Glu387Argfs*14)

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**

Comments
Not detectable in IEF

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

	0.01		

Occurrence comments

gnomAD (2.1)

Overall comments

Occurrence comments

First described in a 52-year-old male with emphysema.

References

Medline ID	Authors	Title
9070606	Brantly M,Lee JH,Hildesheim J,Uhm CS,Prakash UB,Staats BA,Crystal RG,Hildeshiem J	alpha1-antitrypsin gene mutation hot spot associated with the formation of a retained and degraded null variant [corrected; erratum to be published].

Journal	Year	Volume	Num	Pp
American journal of respiratory cell and molecular biology	1997	16	3	225-31

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