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.1177C>T			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 5	M1	Pathogenic
Comments			
rs61761869			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$M_{wurzburg}$		Deficient Precipitating	p.Pro393Ser

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

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
0.64		9195	
Comments			
Variant identified at heterozygous status MwurzburgZ			
Occurrence			
Ethnic background without frequency range :			
Ethnic background and frequency			

Frequency range		Group tested	
from (%)	To (%)	Size	Description (who was tested)
	0.05		

Occurrence comments

Data from gnomAD (2.1)

Overall comments

Occurrence comments

The mutant PI Mwurzburg is completely retained within synthetising cells and the molecular defect of transportation may be similar to that of the common PI Zallele. This variant was identified at heterozygous status MwurzburgZ in a 19-year old man presenting with liver cytolysis. It was also identified at heterozygous status M3Mwurzburg in a 58-year old man presenting with emphysema.

References

Medline ID	Authors	Title
10234508	Poller W,Merklein F,Schneider-Rasp S,Haack A,Fechner H,Wang H,Anagnostopoulos I,Weidinger S	Molecular characterisation of the defective alpha 1-antitrypsin alleles PI Mwurzburg (Pro369Ser), Mheerlen (Pro369Leu), and Q0lisbon (Thr68Ile).

Journal	Year	Volume	Num	Pp
European journal of human genetics : EJHG	1999	7	3	321-31

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