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.275C>T			
Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 2	Unknown	Pathogenic
Comments			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0lisbon}$		Deficient	p.Thr92Ile
3D position of aa affected		Mobility on polyacrylamide gel	Mobility on agarose gel
Near the amino-terminal end in the B alpha-helix			
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
Around 0.94 g/L			
Comments			
This mutant protein is synthetised in quantity similar to that of wild-type allele, but only about one-htird is secreted from the cell.			
Occurrence			
Ethnic background without frequency range : Portuguese			

Ethnic background and frequency			
Frequency range		**Group tested**	
from (%)	To (%)	Size	Description (who was tested)

Occurrence comments

Overall comments
Occurrence comments
Lung disease could occur in homozygotes

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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