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.839A>T;c.710T>C			
Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M1 Ala	Pathogenic
Comments			
rs121912714			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$P_{cardiff}$		Deficient	p.Asp280Val;p.Val237Ala

3D position of aa affected	Mobility on polyacrylamide gel	Mobility on agarose gel
The aa residue at position 280 corresponds to a point where a bêta-sheet strand (B3) undergoes a sharp turn to become an alpha-helix (G)	P	P

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**

Comments
see data for Plowell variant
Occurrence
Ethnic background without frequency range : British

Ethnic background and frequency

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

Occurrence comments

Overall comments

Occurrence comments

see data for Plowell variant

References

Medline ID	Authors	Title
2606478	Graham A,Kalsheker NA,Newton CR,Bamforth FJ,Powell SJ,Markham AF	Molecular characterisation of three alpha-1-antitrypsin deficiency variants: proteinase inhibitor (Pi) nullcardiff (Asp256----Val); PiMmalton (Phe51----deletion) and PiI (Arg39----Cys).

Journal	Year	Volume	Num	Pp
Human genetics	1989	84	1	55-8

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