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

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M1 Val	Pathogenic

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
P_{lowell}		Deficient	p.Asp280Val

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**
0.75		6688	

Comments

The Plowell variant corresponds to the Pcardiff variant with a M1Val genetic background This variant was identified at heterozygous status PlowellS in a 62 year-old smoking man with emphysema. Also his brother received a liver transplant for cirrhosis.

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range		**Group tested**	
from (%)	To (%)	Size	Description (who was tested)
0.01	0.34		

Occurrence comments

from gnomAD (2.1)

Overall comments

Occurrence comments

This variant was also identified at a heterozygous status M1Plowell in a 77-year old man presenting with liver cirrhosis of unknown etiology.

References

Medline ID	**Authors**	**Title**
2240842	Holmes MD,Brantly ML,Crystal RG	Molecular analysis of the heterogeneity among the P-family of alpha-1-antitrypsin alleles.

Journal	**Year**	**Volume**	**Num**	**Pp**
The American review of respiratory disease	1990	142	5	1185-92

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