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.187C>T

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	Unknown	Pathogenic

Comments
rs28931570
AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
I		Deficient Precipitating Dysfunctional	p.Arg63Cys

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

AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	**Homozygous**	**Heterozygous**	**Homozygous**
0.95-1.82		15140	

Comments
May act indirectly on beta-sheet A stability by perturbing helix A/G-core-packing.
Occurrence
Ethnic background without frequency range :

Ethnic background and frequency

Frequency range		Group tested	
from (%)	To (%)	Size	Description (who was tested)
0.30	0.67		

Occurrence comments

Overall comments

Occurrence comments

The polymerization rate of the I variant being very weak, there is no associated hepatic disorder.

References n°1

Medline ID	Authors	Title
10194472	Mahadeva R,Chang WS,Dafforn TR,Oakley DJ,Foreman RC,Calvin J,Wight DG,Lomas DA	Heteropolymerization of S, I, and Z alpha1-antitrypsin and liver cirrhosis.

Journal	Year	Volume	Num	Pp
The Journal of clinical investigation	1999	103	7	999-1006

References n°2

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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