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.374G>A			
Type of variation	Mutation Location	Genetic background	ACMG classification
Neutral SNP	Exon 2	M4	Benign
Comments			
rs709932			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
M_4		Neutral	p.Arg125His

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**

Comments
Normal serum level and anti-elastolytic activity

Occurrence
Ethnic background without frequency range :

Ethnic background and frequency	
Frequency range	**Group tested**

from (%)	To (%)	Size	Description (who was tested)

Occurrence comments

Overall comments

Occurrence comments

This aa substitution defines the M4 genetic background.

Last Update

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