AVB-CG
CENTRE DE RÉFÉRENCE
ATRÉSIE DES VOIES BILIAIRES
ET CHOLESTASES GÉNÉTIQUES

Mutation sequence analysis

Contributed by : CHU de Lille

HGVS nomenclature (NM_000295.4)

Nomenclarure including the signal peptide

c.917+1G>A

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Intron 3	Other	Pathogenic

Comments

genetic background : S

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0achicourt}$		Deficient	No aa change

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

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

Comments

Not detectable in IEF

Occurrence

Ethnic background without frequency range : European

Ethnic background and frequency

Frequency range	**Group tested**

from (%)	To (%)	Size	Description (who was tested)
	0.01		

Occurrence comments

gnomAD(2.1)

Overall comments

Occurrence comments

Described in association with a Q0clayton allele in a 51 year-old man presenting with severe pulmonary emphysema

References

Medline ID	Authors	Title
30223862	Renoux C,Odou MF,Tosato G,Teoli J,Abbou N,Lombard C,Zerimech F,Porchet N,Chapuis Cellier C,Balduyck M,Joly P	Description of 22 new alpha-1 antitrypsin genetic variants.

Journal	Year	Volume	Num	Pp
Orphanet journal of rare diseases	2018	13	1	161

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