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

Mutation sequence analysis			
Contributed by : CHU Lille			
HGVS nomenclature (NM_000295.4)			
Nomenclarure including the signal peptide			
c.552del			
Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 2	M1	Pathogenic
Comments			
rs267606950			

AAT variant and Q0 alleles			
Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
$Q_{0\ \text{Granite falls}}$		Deficient	p.(Tyr184*)

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**
0.62		10938	

Comments
Associated with a S allele.

Occurrence
Ethnic background without frequency range :

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

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

Occurrence comments

no frequency available.

Overall comments

Occurrence comments

Identified at heterozygous status (S Q0granitefalls) in a 73-year old man, presenting with isolated hyperferritinemia (no respiratory or liver troubles)

References n°1

Medline ID	Authors	Title
3040726	Nukiwa T,Takahashi H,Brantly M,Courtney M,Crystal RG	alpha 1-Antitrypsin nullGranite Falls, a nonexpressing alpha 1-antitrypsin gene associated with a frameshift to stop mutation in a coding exon.

Journal	Year	Volume	Num	Pp
The Journal of biological chemistry	1987	262	25	11999-2004

References n°2

Medline ID	Authors	Title
2481421	Holmes M,Curiel D,Brantly M,Crystal RG	Characterization of the intracellular mechanism causing the alpha-1-antitrypsin Nullgranite falls deficiency state.

Journal	Year	Volume	Num	Pp
The American review of respiratory disease	1989	140	6	1662-7

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