

### Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM\_000295.4)

Nomenclature including the signal peptide

c.1130dupT

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 5	M3	Pathogenic

### Comments

This duplication creates a new reading frame from Leu 377 which terminates 23 amino-acids later by a stop codon.

### AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Q <sub>0</sub> mattawa	Q0nancy (Clinica chimica acta 2014, 427:21-22)	Deficient	p.Leu377PhefsX24
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
<b>Heterozygous</b>	<b>Homozygous</b>	<b>Heterozygous</b>	<b>Homozygous</b>
0.68			

### Comments

### Occurrence

Ethnic background without frequency range :

### Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
	0.00			
<b>Occurrence comments</b>				
rs766291631				
<b>Overall comments</b>				
<b>Occurrence comments</b>				
<b>References</b>				
Medline ID	Authors	Title		
2539391	Curiel D,Brantly M,Curiel E,Stier L,Crystal RG	Alpha 1-antitrypsin deficiency caused by the alpha 1-antitrypsin Nullmattawa gene. An insertion mutation rendering the alpha 1-antitrypsin gene incapable of producing alpha 1-antitrypsin.		
Journal	Year	Volume	Num	Pp
The Journal of clinical investigation	1989	83	4	1144-52
<b>Last Update</b>				
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