

### Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM\_000295.4)

Nomenclature including the signal peptide

c.839A>T;c.374 G>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 3	M4	Pathogenic

### Comments

Corresponds to the Pcardiff variant with a M4 genetic background.

### AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine	
P <sub>duarte</sub>		Deficient	p.Asp280Val;p.Arg125His	
3D position of aa affected			Mobility on polyacrylamide gel	Mobility on agarose gel
The aa residue at position 280 corresponds to a point where a bêta-sheet strand (B3) undergoes a sharp turn to become an alpha-helix (G)			P	P
AATserum level (g/L)		Anti-elastolytic activity (IU/L)		
<b>Heterozygous</b>	<b>Homozygous</b>	<b>Heterozygous</b>	<b>Homozygous</b>	

### Comments

see data for Plowell variant

### Occurrence

Ethnic background without frequency range :

<b>Ethnic background and frequency</b>					
<b>Frequency range</b>		<b>Group tested</b>			
from (%)	To (%)	Size	Description (who was tested)		
<b>Occurrence comments</b>					
<b>Overall comments</b>					
<b>Occurrence comments</b>					
see data for Plowell variant					
<b>References n°1</b>					
<b>Medline ID</b>	<b>Authors</b>		<b>Title</b>		
2240842	Holmes MD,Brantly ML,Crystal RG		Molecular analysis of the heterogeneity among the P-family of alpha-1-antitrypsin alleles.		
<b>Journal</b>		<b>Year</b>	<b>Volume</b>	<b>Num</b>	<b>Pp</b>
The American review of respiratory disease		1990	142	5	1185-92
<b>References n°2</b>					
<b>Medline ID</b>	<b>Authors</b>		<b>Title</b>		
8364590	Hildesheim J,Kinsley G,Bissell M,Pierce J,Brantly M		Genetic diversity from a limited repertoire of mutations on different common allelic backgrounds: alpha 1-antitrypsin deficiency variant Pduarte.		
<b>Journal</b>		<b>Year</b>	<b>Volume</b>	<b>Num</b>	<b>Pp</b>
Human mutation		1993	2	3	221-8
<b>Last Update</b>					
First publication : 07-04-2020 22:27 Last update : 08-13-2020 18:11 by Pr Curateur test					