

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.1096G>A

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 5	M1	Pathogenic

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Z		Deficient Precipitating Dysfunctional	p.Glu366Lys
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
Reactive center loop	Z		Z
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
0.58 - 1.1	0.15 - 0.55	11170	3760

Comments

Occurrence

Ethnic background without frequency range : Swedish

Ethnic background and frequency

Frequency range		Group tested		
from (%)	To (%)	Size	Description (who was tested)	
1.00	5.00	12000		
Occurrence comments				
About 1 ZZ individual out of 2500 in France for a global prevalence of about 10,000 ZZ individuals.				
Overall comments				
Occurrence comments				
Most frequent SERPINA1 pathologic variant.				
References				
Medline ID	Authors	Title		
1084290	Jeppsson JO	Amino acid substitution Glu leads to Lys alpha1-antitrypsin PiZ.		
Journal	Year	Volume	Num	Pp
FEBS letters	1976	65	2	195-7
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