

Mutation sequence analysis

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HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.326C>T

Type of variation	Mutation Location	Genetic background	ACMG classification
AAT variant	Exon 2	M1 Val	Pathogenic

Comments

rs199422213

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Z _{bristol}		Deficient	p.Thr109Met
3D position of aa affected	Mobility on polyacrylamide gel		Mobility on agarose gel
			Z Slow
AATserum level (g/L)		Anti-elastolytic activity (IU/L)	
Heterozygous	Homozygous	Heterozygous	Homozygous
0.83		13588	

Comments

associated with a M1 allele.

Occurrence

Ethnic background without frequency range :

Ethnic background and frequency

Frequency range		Group tested			
from (%)	To (%)	Size	Description (who was tested)		
	0.01				
Occurrence comments					
Overall comments					
Occurrence comments					
<p>This variant was identified at an heterozygous status with a M1 allele in a 3-year old girl presenting with frequent bronchitis. This variant was also identified at an heterozygous status with a Z allele in a 47-year old woman presenting with pulmonary emphysema. This variant was also identified at an heterozygous status MZbristol in a 37 year-old man presenting with chronic liver disease with an AAT serum concentration of 0.80 g/L.</p>					
References n°1					
Medline ID	Authors		Title		
9459000	Lovegrove JU,Jeremiah S,Gillett GT, Temple IK,Povey S,Whitehouse DB		A new alpha 1-antitrypsin mutation, Thr-Met 85, (PI Zbristol) associated with novel electrophoretic properties.		
Journal		Year	Volume	Num	Pp
Annals of human genetics		1997	61	Pt 5	385-91
References n°2					
Medline ID	Authors		Title		
23858502	Bates KJ,Puxley M,Hill M,Kalsheker N,Barlow A,Clark BE,Sherwood RA		A patient with the rare alpha-1-antitrypsin variant (Z)bristol in compound heterozygosity with the Z mutation.		
Journal		Year	Volume	Num	Pp
Annals of clinical biochemistry		2013	50	Pt 6	618-21
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