

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.847A>T

Type of variation	Mutation Location	Genetic background	ACMG classification
Null allele	Exon 3	M1 Ala	Pathogenic

Comments

AAT variant and Q0 alleles

Variant name	Also Known as	Pathogenicity	HGVS nomenclature protéine
Q ₀ cairo		Deficient	p.Lys283*
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
			3849

Comments

No band detectable in IEF on agarose gel

Occurrence

Ethnic background without frequency range : Moroccan

Ethnic background and frequency

Frequency range	Group tested

from (%)	To (%)	Size	Description (who was tested)		
0.00	0.00				
Occurrence comments					
Frequency from gnomAD (2.1)					
Overall comments					
Occurrence comments					
Mutation described in a 13 year-old girl presenting with respiratory distress and mild hemoptysis					
References n°1					
Medline ID	Authors		Title		
15905697	Zorzetto M,Ferrarotti I,Campo I,Balestrino A,Nava S,Gorriani M,Scabini R,Mazzola P,Luisetti M		Identification of a novel alpha1-antitrypsin null variant (Q0Cairo).		
Journal			Year	Volume	Num
Diagnostic molecular pathology : the American journal of surgical pathology, part B			2005	14	2
References n°2					
Medline ID	Authors		Title		
29977761	Jouhadi Z,Odou MF,Zerimech F,Bousfiha AA,Mikou N,Porchet N,Crepin M,Najib J,Balduyck M		Alpha1 antitrypsin deficiency due to an homozygous PI* Null Q0Cairo mutation: Early onset of pulmonary manifestations and variability of clinical expression.		
Journal			Year	Volume	Num
Respiratory medicine case reports			2018	24	58-62
Last Update					
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