

Mutation sequence analysis

Contributed by : CHU Lille

HGVS nomenclature (NM_000295.4)

Nomenclature including the signal peptide

c.288_291delTCAC

| Type of variation | Mutation Location | Genetic background | ACMG classification |
|-------------------|-------------------|--------------------|---------------------|
| Null allele | Exon 2 | M2 | Pathogenic |

Comments

AAT variant and Q0 alleles

| Variant name | Also Known as | Pathogenicity | HGVS nomenclature protéine |
|----------------------------|--------------------------------|----------------------------------|----------------------------|
| Q ₀ casablanca | | Deficient | p.His97Metfs*7 |
| 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 |
| | | | 3747 |

Comments

no band detectable on agarose gel Mutation described as Q0lille wich occurs on a PI*Z background

Occurrence

Ethnic background without frequency range : Moroccan

Ethnic background and frequency

| Frequency range | Group tested |
|-----------------|--------------|
|-----------------|--------------|

| from (%) | To (%) | Size | Description (who was tested) | | |
|--|--|------|------------------------------|---|-----|
| | | | | | |
| Occurrence comments | | | | | |
| No frequency reported rs1057516212 | | | | | |
| Overall comments | | | | | |
| Occurrence comments | | | | | |
| Described in a 17 year old patient presenting with severe bronchiectasis | | | | | |
| References | | | | | |
| Medline ID | Authors | | | Title | |
| 30223862 | Renoux C, Odou MF, Tosato G, Teoli J, Abbou N, Lombard C, Zerimech F, Porchet N, Chapuis Cellier C, Balduyck M, Joly P | | | Description of 22 new alpha-1 antitrypsin genetic variants. | |
| Journal | | Year | Volume | Num | Pp |
| Orphanet journal of rare diseases | | 2018 | 13 | 1 | 161 |
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