AVB-CG
CENTRE DE RÉFÉRENCE
ATRÉSIE DES VOIES BILIAIRES
ET CHOLESTASES GÉNÉTIQUES

| **Mutation sequence analysis** | | | |
|---|---|---|---|
| Contributed by : CHU Lyon | | | |
| HGVS nomenclature (NM_000295.4) | | | |
| Nomenclarure including the signal peptide | | | |
| c.1178C>T | | | |
| Type of variation | Mutation Location | Genetic background | ACMG classification |
| AAT variant | Exon 5 | M1 Ala | Pathogenic |
| **Comments** | | | |
| rs199422209 | | | |

| **AAT variant and Q0 alleles** | | | |
|---|---|---|---|
| Variant name | Also Known as | Pathogenicity | HGVS nomenclature protéine |
| $M_{heerlen}$ | | Deficient Precipitating | p.Pro393Leu |

| 3D position of aa affected | Mobility on polyacrylamide gel | Mobility on agarose gel |
|---|---|---|
| | M | M |

| AATserum level (g/L) | | Anti-elastolytic activity (IU/L) | |
|---|---|---|---|
| **Heterozygous** | **Homozygous** | **Heterozygous** | **Homozygous** |
| 0.18 | | 2692 | |

| **Comments** |
|---|
| Variant identified at heterozygous status MheerlenZ |
| **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**

Frequency data from gnomAD (2.1)

**Overall comments**

**Occurrence comments**

The mutant PI Mheerlen is completely retained within synthetising cells and the molecular defect of transportation may be similar to that of the common PI Zallele.

**References**

| Medline ID | Authors | Title |
|---|---|---|
| 10234508 | Poller W,Merklein F,Schneider-Rasp S,Haack A,Fechner H,Wang H,Anagnostopoulos I,Weidinger S | Molecular characterisation of the defective alpha 1-antitrypsin alleles PI Mwurzburg (Pro369Ser), Mheerlen (Pro369Leu), and Q0lisbon (Thr68Ile). |

| Journal | Year | Volume | Num | Pp |
|---|---|---|---|---|
| European journal of human genetics : EJHG | 1999 | 7 | 3 | 321-31 |

**Last Update**

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