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

| **Mutation sequence analysis** | | | |
|---|---|---|---|
| Contributed by : CHU Lille | | | |
| HGVS nomenclature (NM_000295.4) | | | |
| Nomenclarure including the signal peptide | | | |
| c.917+2T>A | | | |
| Type of variation | Mutation Location | Genetic background | ACMG classification |
| AAT variant | Intron 3 | M3 | Uncertain significance |
| **Comments** | | | |
| Mutation located in the 5'splicing site of intron 3. | | | |

| **AAT variant and Q0 alleles** | | | |
|---|---|---|---|
| Variant name | Also Known as | Pathogenicity | HGVS nomenclature protéine |
| $Q_{0lomme}$ | | Unknown | p.* |

| 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** |
| 0.89 | | 12932 | |

| **Comments** |
|---|
| Associated with a M3 allele. |

| **Occurrence** | |
|---|---|
| Ethnic background without frequency range : | |
| **Ethnic background and frequency** | |
| **Frequency range** | **Group tested** |

| from (%) | To (%) | Size | Description (who was tested) |
| --- | --- | --- | --- |
| | | | |

**Occurrence comments**

**Overall comments**

**Occurrence comments**

This variant was identified at a heterozygous status in a 55-year old man presenting with COPD.

**Last Update**

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