

### Mutation sequence analysis

Contributed by : CHU Lyon

HGVS nomenclature (NM\_000295.4)

Nomenclature including the signal peptide

c.514G>A

| Type of variation | Mutation Location | Genetic background | ACMG classification |
|-------------------|-------------------|--------------------|---------------------|
| AAT variant       | Exon 2            | Unknown            | Benign              |

### Comments

rs112030253

### AAT variant and Q0 alleles

| Variant name               | Also Known as                  | Pathogenicity                    | HGVS nomenclature protéine |
|----------------------------|--------------------------------|----------------------------------|----------------------------|
| V                          |                                | Neutral                          | p.Gly148Arg                |
| 3D position of aa affected | Mobility on polyacrylamide gel |                                  | Mobility on agarose gel    |
|                            | V                              |                                  | V                          |
| AATserum level (g/L)       |                                | Anti-elastolytic activity (IU/L) |                            |
| <b>Heterozygous</b>        | <b>Homozygous</b>              | <b>Heterozygous</b>              | <b>Homozygous</b>          |
| 1.07-2.23                  |                                | 22900                            |                            |

### Comments

### Occurrence

Ethnic background without frequency range :

### Ethnic background and frequency

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

|   |   |             |   |            |           |
|---|---|-------------|---|------------|-----------|
| from (%)  | To (%)  | Size        | Description (who was tested)  |            |           |
|   |   |             |   |            |           |
| <b>Occurrence comments</b>  |   |             |   |            |           |
|   |   |             |   |            |           |
| <b>Overall comments</b>   |   |             |   |            |           |
|   |   |             |   |            |           |
| <b>Occurrence comments</b>  |   |             |   |            |           |
|   |   |             |   |            |           |
| <b>References</b>   |   |             |   |            |           |
| <b>Medline ID</b>   | <b>Authors</b>  |             | <b>Title</b>  |            |           |
| 7977369   | Faber JP,Poller W,Weidinger S,Kirchgesser M,Schwaab R,Bidlingmaier F,Olek K |             | Identification and DNA sequence analysis of 15 new alpha 1-antitrypsin variants, including two PI*Q0 alleles and one deficient PI*M allele. |            |           |
| <b>Journal</b>  |   | <b>Year</b> | <b>Volume</b>   | <b>Num</b> | <b>Pp</b> |
| American journal of human genetics  |   | 1994        | 55  | 6          | 1113-21   |
| <b>Last Update</b>  |   |             |   |            |           |
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