

**Supplemental Table 1.** *GLA* gene variants in the cohort

| Amino acid change  |       | Nucleotide change        | Mutation Type  | Likely phenotype*  | n  |
|--------------------|-------|--------------------------|----------------|--------------------|----|
| p.Gly85Asp         | G85D  | c.254G>A                 | Missense       | Classic            | 15 |
| p.Arg112Cys        | R112C | c.334C>T                 | Missense       | Classic            | 10 |
| p.Ala156Thr        | A156T | c.466G>A                 | Missense       | Classic            | 9  |
| p.Ile232Thr        | I232T | c.695T>C                 | Missense       | Likely classic     | 6  |
| p.Asn34Ser         | N34S  | c.101A>G                 | Missense       | Classic            | 4  |
| p.Arg342Ter        | R342X | c.1024C>T                | Nonsense       | Classic            | 3  |
| p.Gly271Ser        | G271S | c.811G>A                 | Missense       | Classic            | 3  |
| p.Arg227Ter        | R227X | c.679C>T                 | Nonsense       | Classic            | 2  |
| p.Tyr329SerfsTer19 |       | c.986delA                | Deletion       | Classic            | 1  |
| p.Gly171Ser        | G171S |                          | Missense       | Not investigated** | 1  |
| p.Arg301Ter        | R301X | c.901C>T                 | Nonsense       | Classic            | 1  |
| p.Asn355Lys        | N355K | c.1065C>A                | Missense       | Classic            | 1  |
|                    |       | c.369+3 c.547+954del4096 | Large deletion | Not investigated** | 1  |

n: number

\* Genotype classification

\*\* There are no available data for the 2 gene variants neither on the International Fabry Disease Genotype-Phenotype Database (dbFGP) (<http://dbfgp.org/dbFgp/fabry/>) nor the <http://fabry-database.org>