**S1 Table. *GALNT12* variants detected in the AAP population. Only c.907G>A (p.D303N) was selected after applying the filtering strategy (red labelled).**

| CHR POSITION | HGVS[a] | rs[b] | ExAC[c] | EVS[d] | 1000G[e] | PD[f] | SA[g] |
|---|---|---|---|---|---|---|---|
| 9:101570336 | c.356A>T; p.E119V | rs10987769 | 0.177 | 0.079 | 0.086 | 2 | NO |
| 9:101585643 | c.477C>G; p.V159= | | 0 | 0 | 0 | na | NO |
| 9:101589058 | c.566A>G; p.N189S | rs183981750 | 0.0007 | 0.0002 | 0 | 0 | NO |
| 9:101594103 | c.781G>A; p.D261N | rs41306504 | 0.0125 | 0.0114 | 0.01 | 2 | NO |
| **9:101594229** | **c.907G>A; p.D303N** | **rs145236923** | **0.0019** | **0.0013** | **0.003** | **2** | **NO** |
| 9:101594263 | c.917+24C>T | rs41297187 | 0.0301 | 0.0229 | 0.019 | na | NO |
| 9:101599421 | c.1203T>G; p.R401= | rs140977555 | 0 | 0 | 0 | na | NO |
| 9:101606425 | c.1392C>G; p.P464= | rs35616709 | 0.0035 | 0.0059 | 0.003 | na | NO |
| 9:101611335 | c.1707G>C p.S569= | rs2273846 | 0.0606 | 0.0521 | 0.069 | na | NO |
| 9:101611545 | c.*171A>G | rs2273847 | - | - | 0.069 | na | na |
| 9:101611697 | c.*323A>C | rs573046827 | - | - | 0.001 | na | na |
| 9:101611795 | c.*421G>A | rs2273848 | - | - | 0.047 | na | na |

[a]HGVS= HGVS variant designation according to the transcript NM_024642 and the protein NP_078918. [b]rs= reference SNP ID. [c]ExAC= MAF for Non-Finnish European population from the Exome Aggregation Consortium database. [d]EVS= MAF for the European-American population from the Exome Variant Server. [e]1000G= MAF for the European population from the 1000 Genomes database. [f]PD= number of programs with protein damage prediction among the three programs tested (SIFT, Polyphen2 and MutationTaster), na=not applicable. [g]SA= splicing alteration prediction according to HSF and MaxEnt algorithms; NO= no alteration prediction, na=not applicable.