**Supplementary Table 1.** ABCA4 variants identified in this cohort.

*Pathogenic* = Truncating alleles, significantly enriched in ABCA4-LOVD; “Likely pathogenic” = Non-truncating alleles, significantly enriched in ABCA4-LOVD; “Unknown pathogenicity” = (AF-ABCA4-LOVD/AF Exac non-Finnish Caucasian) > 1, however not significantly enriched; “Likely benign” = (AF-ABCA4-LOVD/AF Exac non-Finnish Caucasian) < 1; “Benign” = Exac AF > 0.006. AF = allele frequency.

1 These unreported variants either result in a premature stop codon or a frame shift ending in a stop codon. The mRNA produced might be targeted for nonsense mediated decay.

2 The unreported variant c.6287T>C does not alter the protein sequence.

3 The unreported variants c.6287T>C (phospho 9.18 and Grantham: 91) and c.2757A>C (phospho 0.44 and Grantham: 45) are expected to be likely pathogenic and likely benign, respectively.

4 This unreported variant very likely results in a skip of exon 27 resulting in a frameshift.

Several cases were earlier mentioned:


