Supplementary Figure 2. A-D. The LA 3.0 30 Hz amplitudes of patients hemizygous for 15 different mutations, and nullizygous patients, plotted by age. Different categories of mutations based on the analysis of the DA 10.0 a-wave amplitudes are shown separately. The LA 30.0 30 Hz amplitudes were mostly consistent with the classification. E-F. The LA 3.0 30 Hz amplitudes of patients hemizygous or homozygous for the same mutation. Results were mostly consistent with DA 10.0 a-wave analysis (Fig. 4). Mutations p.R212C, p.P1380L and p.R1108C (E) were associated with a notably milder phenotype in homozygous states, suggesting residual ABCA4 function and supporting their categorization as intermediate and not null-like. Conversely, mutations p.E1022K, p.E1087K, p.T1526M and p.C1490Y (F) were associated with similar reduction of amplitudes in hemizygous and homozygous states, with the exception of a patient homozygous for p.T1526M. Dashed lines represent the 95% and 75% confidence intervals of the nullizygous group. The grey area represents the 95% confidence interval of the healthy volunteers.