Supplemental File 10. Amino acid sequences of the wild-type and mutant proteins showing the sequence of the c-terminals (wild-type, blue; mutant, red; aa, amino acid). a) Schematic of the wild-type full length RDI12 protein including the c-terminal amino acid sequence (blue). The unfilled boxes represent the seven coding exons with green boxes representing domains highly conserved in short chain dehydrogenases/reductases, of which RDHs are a subfamily. These domains are the NADP(+) cofactor binding site at amino acids 46-52 (GANTGIG) and catalytic site at amino acids 200-204 (RDVLK).[8] b) Schematic of the mutant RDI12 protein predicted to evade NMD resulting from the c.763delG variant in exon 6. The frameshift results in incorporation of a non-native c-terminus (22 amino acid residues, red) and a premature stop codon leading to a truncated protein.