Supplementary Figure 1 – IQSEC2 mutations in analyzed patients with ID

The coding region of the IQSEC2 gene (NM_001111125.3) in normal individuals (WT). The green and red boxes denote the ATG start codon and the TGA stop codon, respectively. In Patient 1 (PT1) and Patient 2 (PT2), a nucleotide deletion (i.e., pink oval) produced a frameshift with a premature stop codon (i.e., yellow boxes). In Patient 3 (PT3), a guanine insertion (i.e., pink oval) also generated a premature stop codon (i.e., yellow box).