Table S1, available as an excel file, show variant calling statistics for next-generation exome sequencing in kindred A.

Table S2, available as an excel file, shows the gene expression profile of Th1-, Th2-, and Th17-associated cytokines in IL-21R-deficient patient P2 of family A.

Table S3, available as an excel file, shows the primer for Sanger sequencing of IL21R and RAB3IL1.