Guide to Thaventhiran *et al.* Supplementary Files

File: Supplementary Information.pdf

This document contains: Strategy Outline that provides the rationale for the prioritisation approaches undertaken within the study; Supplementary Methods that provide additional recruitment and ethics details; and five Supplementary Notes clarifying clinical information and providing further details of some of the analytical strategies described in the manuscript.

File: Supplementary Table 1 – Diagnostic findings.docx

This table contains the list of PID genes and clinically relevant variants identified in the NBR-PID cohort, phenotypes of the patients with reported variants, and OMIM phenotypes associated with defects in the same genes.

File: Supplementary Table 2 – BeviMed genes and variants.xlsx

This table is a comprehensively annotated list of variants that contribute to the BeviMed prioritisation of candidate genes as causes of PID.

File: Supplementary Table 3 – candidate cHET.xlsx

This table lists candidate pathogenic non-coding deletions of a conserved regulatory element (CRE) of a known PID gene, in which the same patient also has a rare damaging exonic variant. Compound heterozygosity (cHET) for a non-coding CRE deletion and a coding variant could lead to effectively absent gene expression and a recessively inherited disease.

File: Supplementary Table 4 – COGS genes.xlsx

This table lists candidate PID genes within the AD-PID GWAS loci, prioritised through capture-HiC omnibus gene score (COGS) approach.

File: Supplementary Figure 1 – raw blots.pdf

This file contains raw images of Western blots, and gating strategies for B and T cell immunophenotyping experiments.