### Supplementary Table 2: A primary immunodeficiencies gene panel covering 499 genes revealed no likely pathogenic variants.

**A: Identified variants with whole-exome sequencing.**

| Chr | Start | Stop | Transcript | Gene | HGVS cDNA-level | HGVS protein-level | dbsNP | gnomAD AF | gnomAD MAF | Zygosity | Classification |
|-----|-------|------|------------|------|----------------|-------------------|-------|-----------|------------|----------|----------------|
| chr1 | 154436093 | 154436094 | NM_000565.4 | IL6R | c.932T>C | p.(Met311Thr) | rs75508163 | 0.00002093 | 0.00009579 | heterozygous | CLASS 3 |
| chr1 | 198752693 | 198752694 | NM_002838.5 | PTTPC | c.320G>A | p.(Val107Ile) | rs139014596 | 0.000174 | 0.0001684 | heterozygous | CLASS 3 |
| chr1 | 235759071 | 235759072 | NM_000081.3 | LYST | c.6782G>A | p.(Arg226His) | rs147791378 | 0.0001365 | 0.00095 | heterozygous | CLASS 2 |
| chr4 | 56478617 | 56478618 | NM_069947.4 | SRPT2 | c.793G>A | p.(Val265Ile) | rs756512943 | 0.00000796 | 0.0003027 | heterozygous | CLASS 2 |
| chr5 | 66053675 | 66053676 | NM_01253697.1 | ERBIN | c.2357C>T | p.(Thr786Ile) | rs752498860 | N/A | N/A | heterozygous | CLASS 3 |
| chr6 | 39134271 | 39134272 | NM_00063.6 | C2 | c.821G>A | p.(Lys274Arg) | N/A | N/A | heterozygous | CLASS 3 |
| chr7 | 117531068 | 117531069 | NM_00492.3 | CFTR | c.443C>T | p.(Ile148Thr) | rs35516286 | 0.0001671 | 0.000694 | heterozygous | CLASS 3 |
| chr8 | 99135045 | 99135046 | NM_017890.4 | VPS13B | c.1333T>G | p.(Cys445Gly) | N/A | N/A | heterozygous | CLASS 3 |
| chr9 | 428369 | 428370 | NM_2034473 | DOCK8 | c.4364G>T | p.(Ser1449Leu) | rs370123223 | 0.00005585 | 0.0001395 | heterozygous | CLASS 3 |
| chr11 | 154436093 | 154436094 | NM_000565.4 | IL6R | c.932T>C | p.(Met311Thr) | rs75508163 | 0.00002093 | 0.00009579 | heterozygous | CLASS 3 |
| chr11 | 198752693 | 198752694 | NM_002838.5 | PTTPC | c.320G>A | p.(Val107Ile) | rs139014596 | 0.000174 | 0.0001684 | heterozygous | CLASS 3 |
| chr12 | 43777662 | 43777663 | NM_016123.3 | IRAK4 | c.749G>A | p.(Gly250Asp) | rs769470855 | 0.0000426 | 0.0002786 | heterozygous | CLASS 2 |
| chr13 | 42606498 | 42606499 | NM_033012.3 | TNFSF11 | c.315T>C | p.(Gly105=) | rs146464645 | 0.00002302 | 0.0001504 | heterozygous | CLASS 2 |

The nomenclature of the identified variants is according to the HGVS guidelines (http://www.hgvs.org) with nucleotide 'A' of the ATG start codon = 'c.1'. The position of the reported variants is based on NCBI build GRCh38.

### B: Gene list of 499 sequenced genes in the primary immunodeficiencies gene panel.

| Chr | Start | Stop | Transcript | Gene |
|-----|-------|------|------------|------|
| chr1 | 154436093 | 154436094 | NM_000565.4 | IL6R |
| chr1 | 198752693 | 198752694 | NM_002838.5 | PTTPC |
| chr1 | 235759071 | 235759072 | NM_000081.3 | LYST |
| chr4 | 56478617 | 56478618 | NM_069947.4 | SRPT2 |
| chr5 | 66053675 | 66053676 | NM_01253697.1 | ERBIN |
| chr6 | 39134271 | 39134272 | NM_00063.6 | C2 |
| chr7 | 117531068 | 117531069 | NM_00492.3 | CFTR |
| chr8 | 99135045 | 99135046 | NM_017890.4 | VPS13B |
| chr9 | 428369 | 428370 | NM_2034473 | DOCK8 |
| chr11 | 154436093 | 154436094 | NM_000565.4 | IL6R |
| chr11 | 198752693 | 198752694 | NM_002838.5 | PTTPC |
| chr12 | 43777662 | 43777663 | NM_016123.3 | IRAK4 |
| chr13 | 42606498 | 42606499 | NM_033012.3 | TNFSF11 |

The position of the reported variants is based on NCBI build GRCh38.