Supplemental material

Zhang et al., https://doi.org/10.1084/jem.20182304

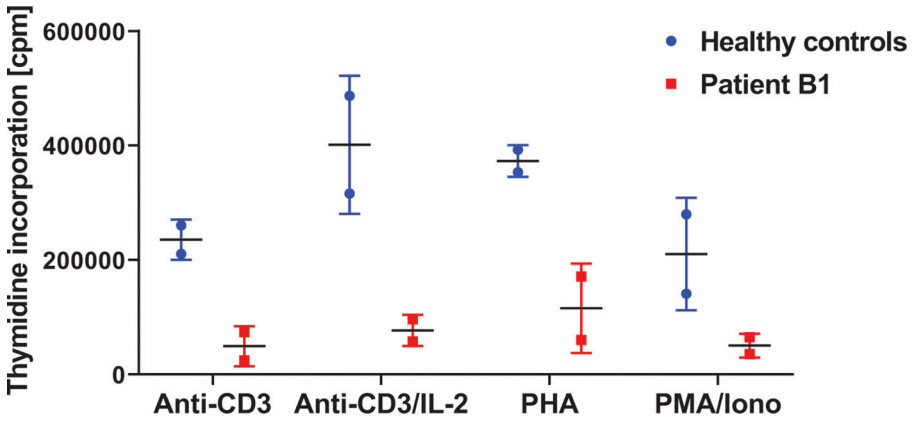

Figure S1. **T cell–proliferative responses. Graph of T cell–proliferative responses to PHA, anti-CD3, anti-CD3, and IL-2, and PMA/ionomycin (Iono) in patient B1 and healthy control cells based on thymidine incorporation.**

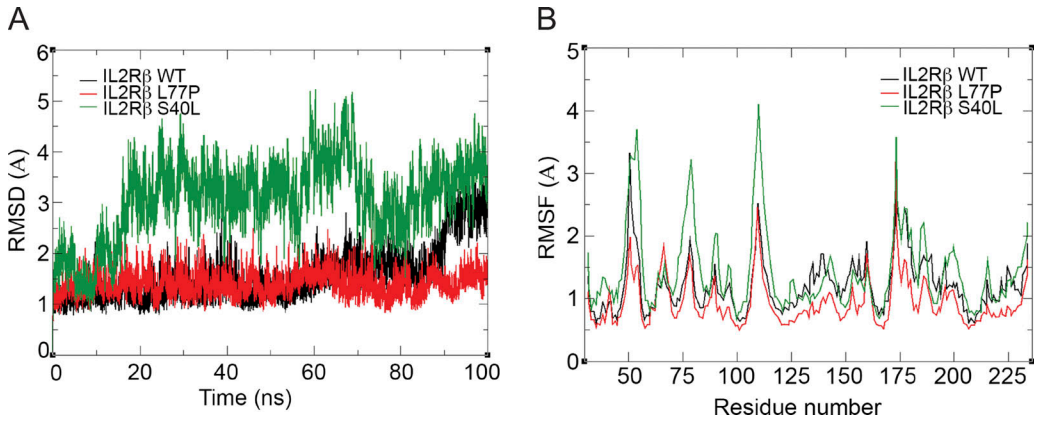

Figure S2. **Molecular dynamic simulation of WT, S40L, and L77P IL-2Rβ structures. (A)** Root mean square deviation (RMSD) between the WT, S40L, and L77P IL-2Rβ structures over 100 ns of unrestrained MD simulation with explicit solvent. **(B)** Root mean square fluctuation (RMSF) in WT, S40L, and L77P IL-2Rβ over the complete trajectory.

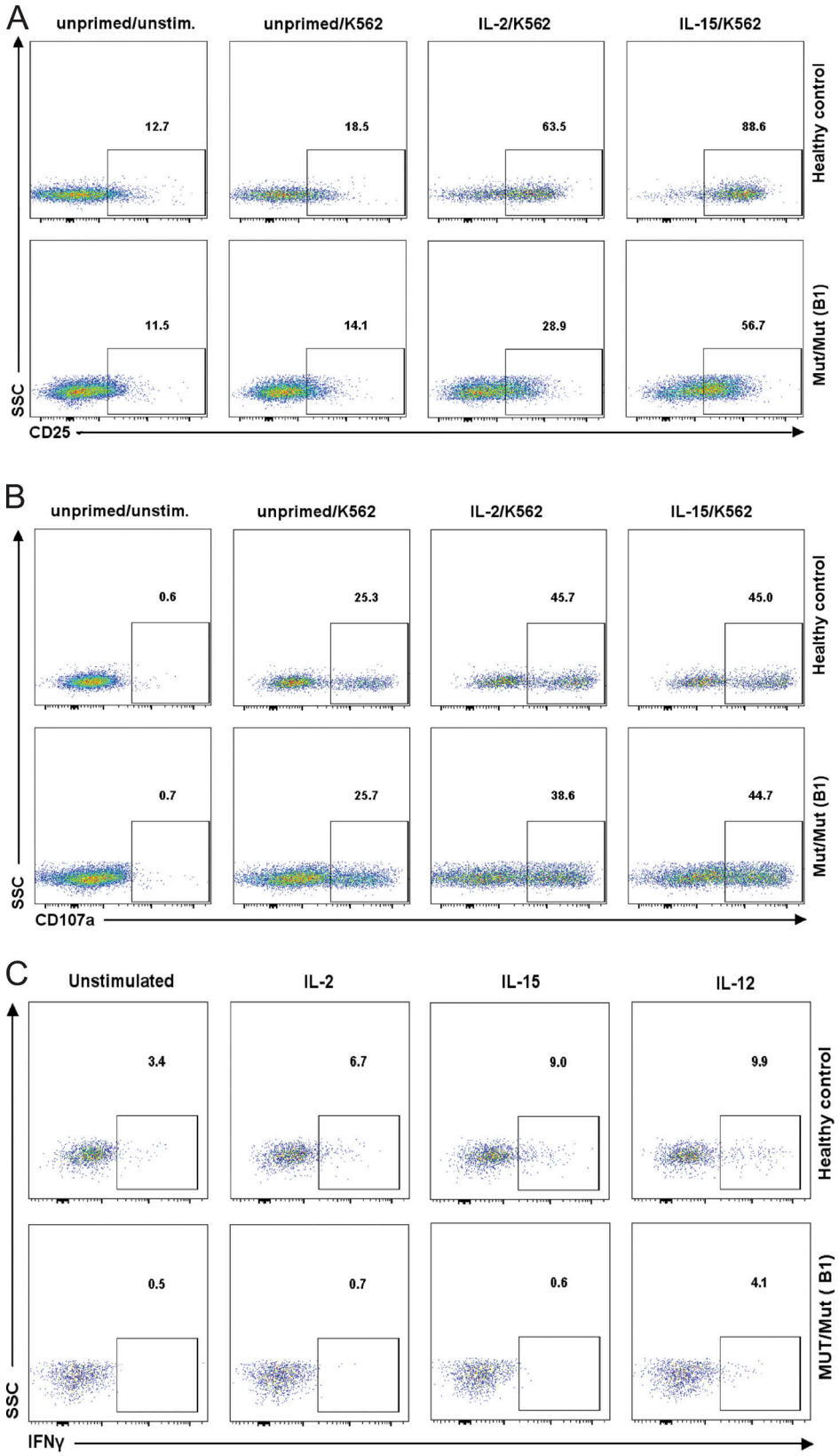

Figure S3. **NK cell immune phenotype. (A)** FACS plots demonstrating up-regulation of CD25 in $CD3^{-}$ $CD56^{+}$ NK cells from a healthy control (top panel) or patient B1 (bottom panel) either unprimed or after priming with IL-2 or IL-15 (100 ng/ml each) for 12 h followed by a 3 h coincubation period with K562 cells. SSC, side scatter. **(B)** FACS plots showing surface expression of CD107a in CD3-$CD56^{+}$ NK cells from a healthy control (top panel) or patient B1 (bottom panel) either unprimed or after priming with IL-2 or IL-15 (100 ng/ml each) for 12 h in response to coincubation period K562 cells for 3 h. **(C)** Intracellular staining for IFNγ in $CD3^{-}$ $CD56^{+}$ NK cells from a healthy control (top panel) or patient B1 (bottom panel) after stimulation with IL-2, IL-15, or IL-12 (100 ng/ml each) for 6 h.

Tables S1, S2, and S3 are provided online as separate Excel files. Table S1 lists patient mutations and clinical manifestations. Table S2 lists lab values and absolute cell counts. Table S3 lists rare variants that were identified and cosegregated in each kindred.