Figure S1: Characterization of studied patients and controls

(A) ALPS subtype, number and age (in years) of studied patients and controls. (B) Percentage of circulating DNT (CD4-CD8- of CD3+TCRαβ+ T-cells), closed circles: ALPS-FAS, open circles: ALPS-sFAS, squares: ALPS-FAS-sLOH, triangles: adults controls.
Figure S2: ALPS patients have increased CD4low and CD8low T-cells resembling DNT

(A) Representative plots showing increased CD8low T-cells in one patient (P1) and a pronounced increase of CD4low T-cells in another ALPS-FAS-sLOH patient (P2). (B) Percentages of CD4low and CD8low T-cells were compared between all ALPS patients (ALPS all) and adult controls (Con>18y); (ns: not significant, * p<0.05, ** p< 0.01, *** p<0.001). (C) The phenotype of T-cells of a representative ALPS-FAS patient is shown for CD4+, CD4low, DNT, CD8low and CD8+ T-cells as defined by the regions in the upper dot plot.
Figure S3: Reduced Fas expression on DNT of patients with ALPS-FAS-sLOH

(A) Fas expression on CD4+, CD8+ and DNT-cells of a patient with ALPS-FAS-sLOH versus a healthy control donor. (B) Percentage of mutated allele of CD4+, CD8+ and DNT-cells in the same patient. The arrow points to the size of the peak of the mutated nucleotide.
Figure S4: Double mutated T-cells accumulate among CD4+ CCR7-CD45RA+ T-cells in patients with ALPS-FAS-sLOH

Fas expression is shown on naïve (naïve, CCR7+CD45RA+), central memory (CM, CCR7+CD45RA-), effector memory (EM, CCR7-CD45RA-) and terminally differentiated CD4+ T-cells (TEMRA, CCR7-CD45RA+) of a 32 year-old patient with ALPS-FAS-sLOH versus an adult control. Control CD4+TEMRA were not detectable (n.det.).
Figure S5: DNT of patients with ALPS-like diseases differ from DNT of FAS mutant individuals

(A) The percentage of circulating DNT (CD4-CD8- of CD3+TCRαβ+ T-cells) is shown for ALPS patients (closed circles: ALPS-FAS, open circles: ALPS-sFAS, squares: ALPS-FAS-sLOH), for adults controls (triangles) and for patients with ALPS-like diseases (closed diamonds: ALPS phenotype with lymphoproliferation and autoimmune cytopenia(s), open diamonds: CVID, mixed diamond: SLE). (B) Percentages of CCR7-CD45RA+TEMRA among DNT and percentages of DNT expressing CD57, CD127 or KLRG1 were compared between ALPS patients, controls and patients with ALPS-like diseases (Disease Controls); (ns: not significant, * p<0.05, ** p< 0.01, *** p<0.001).