Figure S1. Flow cytometric analysis of peripheral blood lymphocytes from controls and patients with mutations in Btk or BLNK
Cells were stained with FITC labeled polyclonal goat anti-IgM and PE labeled CD19. For the control samples, approximately 20,000 events were analyzed; for the patient samples, between 100,000 and 500,000 events were analyzed. The specific mutations in Btk were A582V, L295V and Q157X (from top to bottom). The specific mutation in BLNK was a homozygous R123X.
Figure S2. Expression of CD19 on transitional B cells from a healthy control and a patient with a mutation in Btk (Q157X). Cells were stained with APC labeled CD19, FITC labeled CD38 and PE labeled CD24. The CD19 positive cells, shown on the top, were analyzed for expression of CD38 and CD24 (bottom). The mean fluorescence intensity of the total CD19 population and the CD38/CD24 bright transitional cells are shown in the upper right corner of each plot.
Figure S3. Reduced expression of CD34 on B cell precursors from Pt. 2 and Pt. 3 compared to a healthy control and disease controls

Bone marrow cells from a healthy child (top left), patients with a C506F mutation in Btk (top middle), a homozygous alteration in the alternative splice site of μ heavy chain (top right), a heterozygous deletion of the μ heavy chain locus on one allele with a 2 bp deletion in exon 2 of μ on the other allele (bottom left), Pt. 2 (bottom middle) and Pt. 3 (bottom right) were stained with antibodies to CD19 and CD34 then permeabilized and stained with antibodies to cytoplasmic CD179a (VpreB) and TdT. Expression of CD34 and CD179a was analyzed on all cells positive for CD19 and/or TdT.