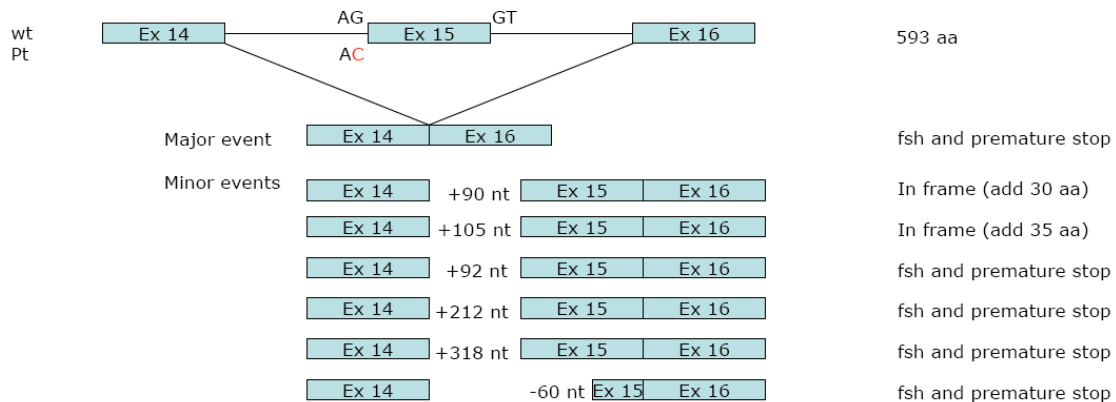


**Supplemental Data****AJHG, Volume 85****Familial Hemophagocytic Lymphohistiocytosis Type 5****(FHL-5) Is Caused by Mutations in Munc18-2****and Impaired Binding to Syntaxin 11**

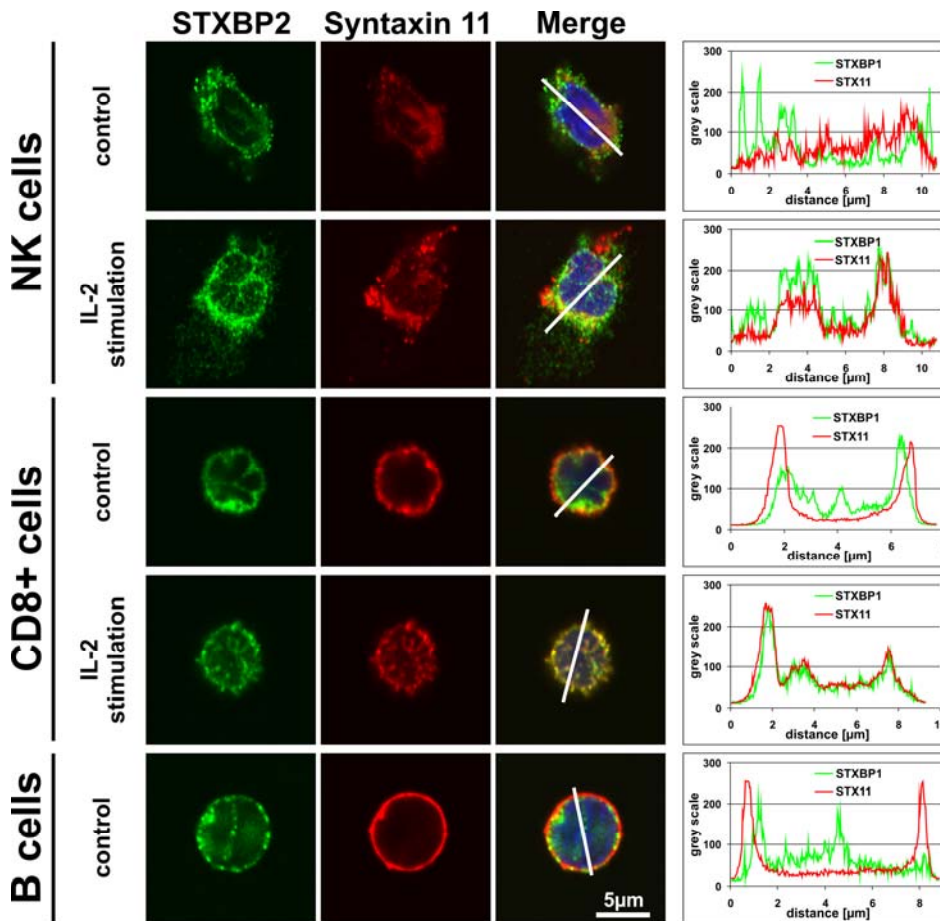
Udo zur Stadt, Jan Rohr, Wenke Seifert, Florian Koch, Samantha Grieve, Julia Pagel, Julia Strauß, Brigitte Kasper, Gudrun Nürnberg, Christian Becker, Andrea Maul-Pavicic, Karin Beutel, Gritta Janka, Gillian Griffiths, Stephan Ehl, and Hans Christian Hennies

**Figure S1. Effects of the Splice Site Mutation c.1247-1G>C in Patient 0879**



Total RNA was analysed by reverse transcription of a fragment including exons 14 to 16. PCR analysis of the cDNA showed a variety of products. Fragments were cloned and single clones were sequenced. A complete loss of exon 15 was found in about 95% of the clones, resulting in a frameshift and a stop codon after 126 new codons and a potential protein of 541 residues. Two minor in-frame products were identified, all further products showed a frameshift (fsh) and an early stop codon, probably resulting in instable mRNA.

Figure S2. Cellular Localization of STXBP2 and Syntaxin 11



Primary CD8<sup>+</sup> T and NK cells showed a clear co-localization of STXBP2 and syntaxin 11, which was more pronounced in cells stimulated with IL-2, showing twice as many cells with a co-localization than without stimulation. There was a broader distribution of STXBP2 in B cells and a weaker co-localization with syntaxin 11.

**Table S1. PCR Primers Used for the Exon Amplification**

Stxbp2_ex1F	TCC ACG TGG GTG ACG CG
Stxbp2_ex1R	TGA AAC CCA GGA TCC CAG AG
Stxbp2_ex2F	ACT TTA GCC TAG GTG ACA GAG TG
Stxbp2_ex2R	ACA TTC AGG GTC TTC CCC ATA G
Stxbp2_ex3F	AGC CAG CCC TTG AAA CTG C
Stxbp2_ex3R	TCC TAC ACT CAA GGG CTG AAC
Stxbp2_ex4F	AGT GGG TTT CTC CTG CAG TC
Stxbp2_ex4R	ATG TAC ACG CAT GCA CGC G
Stxbp2_ex5F	TTG CAC ATG GTG GCA GAT GG
Stxbp2_ex5R	ATC AGG CCA GAG CAG CCC
Stxbp2_ex6F	AGA GCC TAG GGT GTT GGT G
Stxbp2_ex6R	ACA GGT ATG AGC CAC TGC G
Stxbp2_ex7F	AGG TGT GCA CCT GCA GCG
Stxbp2_ex7R	AGA GGT TAA AGG TTA GGT GTT GC
Stxbp2_ex8F	TGA CTC TCA CCT TCA AAC CCA T
Stxbp2_ex8R	AGG AAT GAG GCA CTG ACC CT
Stxbp2_ex9F	TTC AAG GCA GAC ACT CCC AG
Stxbp2_ex9R	ACT GGG GAA CCA GGT CAG T
Stxbp2_ex10F	AGA GCA GGA CAC ATA CAG GTC
Stxbp2_ex10R	AAG ACT TGG CAG CAG CCA G
Stxbp2_ex11F	TGG GCT CAC CCA TGG CCT
Stxbp2_ex11R	TCT CAG TCC AGG CTC GAA ATC
Stxbp2_ex12F	TTC GAG CCT GGA CTG AGA C
Stxbp2_ex12R	AGG CTG AGT CAA GGC AGA AC
Stxbp2_ex13F	ATG CTC TGT CTG CGT TCT GC
Stxbp2_ex13R	TCT CTA CAG TTC AGC CCC TC
Stxbp2_ex14F	TGA GGT AGG ACC CAA ATG TCC
Stxbp2_ex14R	TCA GCA GTT CCC AGG ATC GT
Stxbp2_ex15F	TCT CTC ATC AGA GTC CTG GG
Stxbp2_ex15R	AGG AAC CAC ACC GTG CCT G
Stxbp2_ex16F	TAC CAG CCA CAC CAG GCG
Stxbp2_ex16R	ATG CCA GCA ATG AGC ATT TTC C
Stxbp2_ex17F	TGT CTG CCA GGA TCT TGG C
Stxbp2_ex17R	TAA CAC AGA CCC AGG CGT G
Stxbp2_ex18F	ACC TGT GGC CCT TCG TAT C
Stxbp2_ex18R	ACT CTG GAG TCC AGA CAC C
Stxbp2_ex19F	AGC CTG TCA AAG ACG AAG GC
Stxbp2_ex19R	AGA AGG TGA GGT AGG CAC TG

**Table S2. Sequences of qPCR Primers Used**

Description	Forward primer	Reverse primer	Product
<i>GAPDH</i>	TGCACCACCAACTGCTTAGC	GGCATGGACTGAGGTCATGAG	87 bp
<i>ACTB</i>	AGCCTCGCCTTTGCCGA	CTGGTGCCTGGGGCG	174 bp
<i>TFRC</i>	GGATAAAGCGTTCTTGGTACCA	CCAGTAACCGGATGCTTCACA	105 bp
18S rRNA	CATGCTAACTAGTTACGCGACC	CATCACAGACCTGTTATTGCTC	106 bp
<i>STXBP2</i>	CTTCCTGCTGCAAAATGTCA	GCAAATAAATGGCCTCCAGA	105 bp
<i>STX11</i>	ACAGGTTTCCTTCTCCATCG	TGCTGGTCATATTGCTTGGA	159 bp