

Online Materials for

Munc18-2/STXBP2 deficiency causes familial hemophagocytic lymphohistiocytosis type 5 (FHL5) and impairs cytotoxic granule exocytosis.

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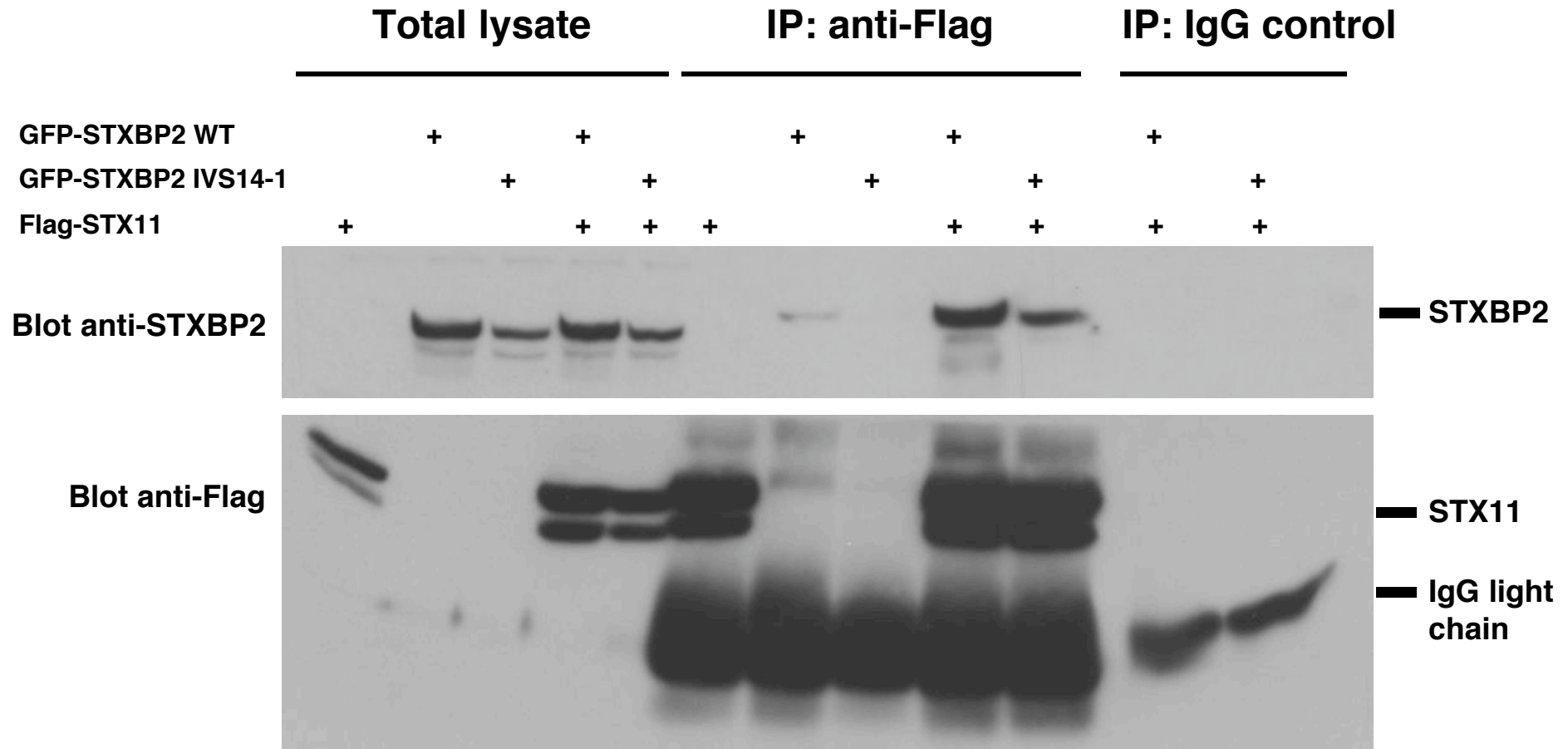
Supplementary Table 1. List of oligonucleotides

	Oligonucleotide annotation	Sequence 5'....3'
Primers for cDNA analyses	STXBP2 Forward	AGAAGGATGGGGAGTGGAAG
	STXBP2 Reverse	CGGAAGGGGCAGTAGAGGT
	Actin Forward	TACCACTGGCATCGTGATGGACT
	Actin Reverse	TCCTTCTGCATCCTGTCCGGCAAT
Primers for genomic DNA analyses	Exon 1 Forward	ACTCCCATCTGTGAGCGTGT
	Exon 1 Reverse	AACCCAGGATCCCAGAGC
	Exons 2, 3 Forward	GGGAATTAAGATGGGGGTTTC
	Exons 2, 3 Reverse	GGGAGTTCTGGACGATTTGA
	Exon 4 Forward	GTCCCAGTCACTTTGCCATT
	Exon 4 Reverse	ACATGGACACATGCACACG
	Exons 5, 6 Forward	GTTTGCACATGGTGGCAGAT
	Exons 5, 6 Reverse	CCCAGCCTCA GTGTCTGTTT
	Exon 7 Forward	AGGCATGGGTTTAGGGTTG
	Exon 7 Reverse	CTGGGGTCACAAGAGAGGTT
	Exons 8, 9, 10 Forward	TCTCACCTTCAAACCCATCC
	Exons 8, 9, 10 Reverse	TCATGAGGGCCAAAGACTTG
	Exons 11, 12, 13 Forward	AGACTGGCCTCCAATTTAC
	Exons 11, 12, 13 Reverse	CCCCCATCTCTACAGTTCAG
	Exons 14, 15 Forward	TCCCCTTCCCTGACACATAG
	Exons 14, 15 Reverse	GAACCACACCGTGCCTGT
Exon 16 Forward	CCCTTTGGCTTTCTCTCTCC	
Exon 16 Reverse	CTGTCACCCCAATTCCTCAG	
Exons 17, 18, 19 Forward	GTGGAGATGCTGGTTCCTGT	
Exons 17, 18, 19 Reverse	TTAACCTCCATGAGGGCTTG	
Primers for cloning	STXBP2 Forward	GAATTCTATGGCGCCCTCGGGG
	STXBP2 Reverse	GGATCCTCAGGGCAGGGCAATG
	STX11 Forward	CGGAATTCCGGTGGTGCAGGCA AAATGAAAGAC
	STX11 Reverse	CCCTCGAGGGCGGGATCCCGGC

Supplementary Table 2. List of antibodies and labeled proteins

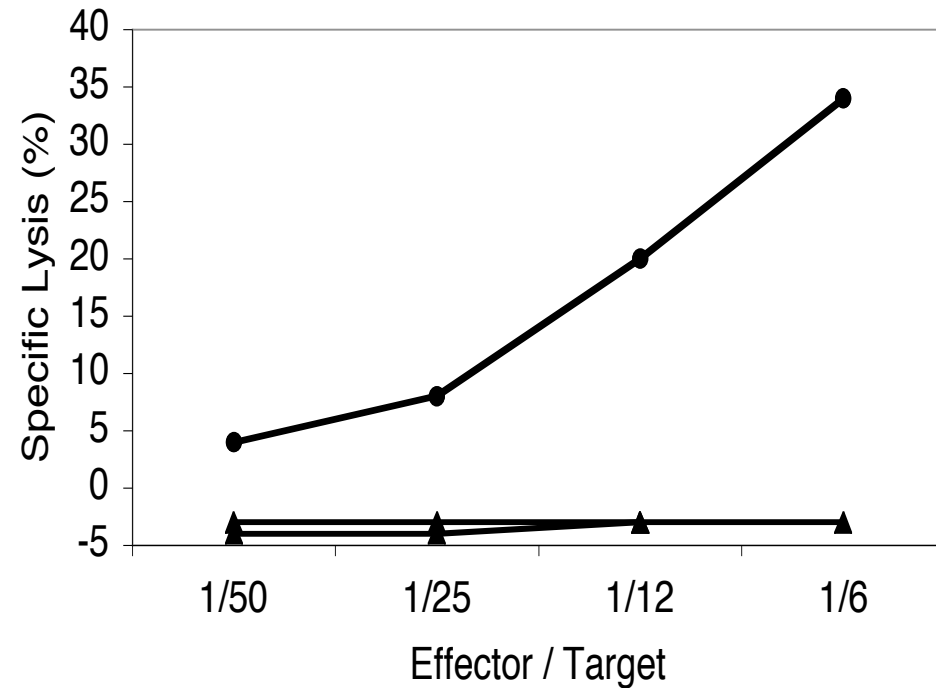
Antigen	Labeling	Manufacturer	Catalogue No.
CD107a	FITC	BD Biosciences	555800
CD107b	FITC	BD Biosciences	555804
CD3	APC	BD Biosciences	345767
CD56	PE	BD Biosciences	555516
CD8	PE	BD Biosciences	555867
CD16	-	Beckman Coulter	IM0813
perforin	Alexa Fluor® 488	BioLegend	308108
PI3K	-	Upstate	06-195
syntaxin 3	-	Sigma	S5547
FLAG	-	Sigma	F3165
GFP	-	Roche	11 814 460 001
anti-mouse	HRP	GE Healthcare Amersham	NA9310V
protein-A	HRP	GE Healthcare Amersham	NA9120V
phalloidin	Alexa Fluor® 594	Invitrogen	A-12381

Supplementary Figure S1



Supplementary Figure 1. STX11 and STXBP2 (wild-type and splice mutant) interact in transfected 293T cells. A FLAG-tagged STX11 and a GFP-tagged STXBP2-wt or -IVS14-1 mutant were cotransfected in 293T cells. Immunoprecipitation was carried out with FLAG-specific antibody or IgG control, and Western blotting with FLAG-, or STXBP2-specific antibodies. Data are representative of four independent experiments.

Supplementary Figure S2



Supplementary Figure 2. Defective cytotoxic activity of STXBP2-deficient NK cells. The cytotoxic activity of resting NK cells from control (circle) and STXBP2-deficient patients (F5P1 and F5P2, triangle) was measured at different effector-K562 target ratio by using a standard ^{51}Cr release assay.