CCL22 mutations in large granular lymphocytic leukemia

Authors

Yuga Mizuno,[1*] Toru Kawakami,[2*] Daigo Higano,[1] Shotaro Miyairi,[1] Ami Asakura,[1] Fumihiro Kawakami,[2] Keijiro Sato,[3] Shuji Matsuzawa,[2] Sayaka Nishina,[2] Hitoshi Sakai,[2] Yumiko Higuchi,[1,4] Kazuyuki Matsuda,[1,5] Hideyuki Nakazawa[2] and Fumihiro Ishida[1,2,4]

[1]Department of Clinical Laboratory Investigation, Graduate School of Medicine, Shinshu University, Matsumoto; [2]Department of Hematology, Shinshu University School of Medicine, Matsumoto; [3]Department of Hematology, Nagano Red Cross Hospital, Nagano; [4]Department of Biomedical Laboratory Sciences, Shinshu University School of Medicine, Matsumoto and [5]Department of Health and Medical Sciences, Graduate School of Medicine, Shinshu University, Matsumoto, Japan

*YM and TK contributed equally as first authors.

Correspondence:
F. ISHIDA - fumishi@shinshu-u.ac.jp

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Supplemental Materials

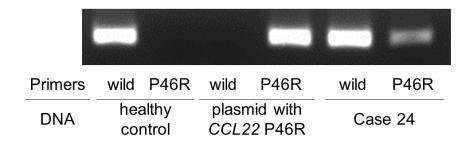Supplement to: Mizuno Y, et al. *CCL22* mutations in large granular lymphocytic

leukemia

Contents:

Supplemental Figure S1.

Primers	wild	P46R	wild	P46R	wild	P46R
DNA	healthy control		plasmid with *CCL22* P46R		Case 24	

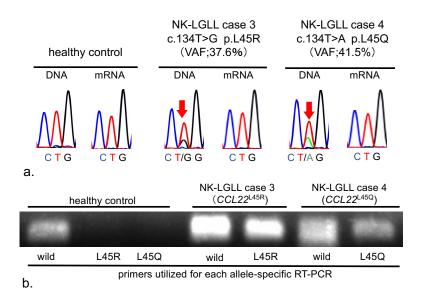**Allele-specific polymerase chain reaction (PCR) for *CCL22* P46R mutation.**

Case 24 was a patient with CD8[+] TCRαβ type T cell large granular lymphocytic (T-

LGL) leukemia. Allele-specific PCR was performed using primers for wild-type

CCL22 or *CCL22* P46R mutation, which validated a *CCL22* P46R mutation identified

with the target sequencing. *CCL22* P46R mutation was also confirmed in case 25 with

the same method. Primer sequences would be provided upon proper requests to the

corresponding author.

Supplemental Figure S2.

a: Sanger sequencing analyses showing healthy control, NK-LGLL case 3 (c.134T>G p.L45R, VAF;37.6%), and NK-LGLL case 4 (c.134T>A p.L45Q, VAF;41.5%) with DNA and mRNA traces.

b: Allele-specific RT-PCR analyses showing healthy control, NK-LGLL case 3 (CCL22^L45R), and NK-LGLL case 4 (CCL22^L45Q). Primers utilized for each allele-specific RT-PCR.

The CCL22 mRNA expression in peripheral blood cells from patients with NK cell large granular lymphocytic leukemia.

a: Sanger sequencing analyses of a *CCL22* mutational site in NK-LGLL patients with *CCL22* mutations. Red arrow indicates a heterozygous C. 134T>G p.L45R or C. 134T>A. L45Q mutation in case 3 or 4, respectively, that was identified in amplified DNA from genome DNA derived from peripheral blood mononuclear cells (PBMC), while no mutational peak was recognized in cDNA reverse-transcribed from mRNA of PBMC.

b: Allele-specific RT-PCR analyses of *CCL22* mutational sites in NK-LGLL patients with *CCL22* L45 mutation.

Supplemental Table S1: NK-LGL leukemia patients with *CCL22* mutations.

case No.	Age (y.o.)	Sex	Neu (x10⁹/L)	Lym (x10⁹/L)	Hb (g/dL)	PLT (x10⁹/L)	*CCL22* mutations (VAF%)	complication	therapy	observation periods (years)	outcome
3	57	F	3.2	8.6	12.6	280	L45R(37.6)	neuropathy	Watch	3	alive
4	73	F	3.0	6.2	12.0	274	L45Q(41.5)	none	Watch	9	alive
5	82	M	3.9	7.3	15.9	286	L45R(31.8)	oculomotor palsy	Watch	2	alive
6	80	M	3.4	8.6	15.6	121	L45Q (9.9)	NAFLD	Watch	0.2	alive
7	56	M	2.5	6.4	14.3	231	L45Q(34.5)	none	Watch	1	alive
8	28	F	0.6	1.1	11.7	286	L45R(29.7)	SCPTCL	PSL	4	alive

NAFLD, non alcoholic fatty liver disease ; SCPTCL, subcutaneous panniculitis-like T

cell lymphoma; PSL, prednisolone