SUPPLEMENTAL MATERIAL

Pachlopnik Schmid et al., http://www.jem.org/cgi/content/full/jem.20121303/DC1

Table S1. Immunological features of FILS cases

Parameter	Patient ID															
	Group 1			Group 2						Group 3						
	VI-28	VI-39	Normal values	VII-2	VI-38	VI-29	VII-1	VI-36	Normal values	V-1	VI-31	VI-3	VI-10	VI-12	VI-11	Normal values
Age (yr)	3	6	2–6	6	8	10	11	12	6–12	17	17	23	24	31	33	12–adult
Ig level (mg/ml)																
IgM	0.20	0.16	0.4–1.0	0.44	0.19	0.29	0.26	0.30	0.6–2.1	0.22	0.08	0.32	0.23	0.34	0.44	0.6–2.1
IgG	5.3	6.2	3.5–11.8	11.1	9.1	11.4	7.82	13	4.8–14.0	12.5	N	10.0	5	6.5	13.3	5.2–12.3
IgA	0.2	0.89	0.4–1.1	1.3	0.97	2.27	1.62	0.37	0.7–1.9	1.95	3.95	1.43	0.76	0.87	2.70	1.16–2.6
Allohemagglutinin titer	1:1	1:1	>1:8	1:8	1:4	1:8	nd	1:4	>1:16	1:1	1:1	nd	nd	nd	1:4	>1:16
Antibodies after immunization																
Anti–*S. pneumoniae* polysaccharide IgG (µg/ml)	0.8[a]	1.0[a]	>7	2.3[a]	0.5[a]	nd	1	0.5[a]	>7	2.4[a]	0.3[a]	nd	nd	nd	nd	>7
Anti-polio (neutralizing antibodies against poliovirus types 1/2/3)	>160	nd	>5	40/160/60	80/160/160	60/80/160	60/160/5	nd	>5	nd	20/60/20	nd	nd	nd	120/60/10	>5
Anti–tetanus toxoid (UI/ml)	2.5	nd	>5	0.11[a]	0.54	0.60	<0.1[a]	nd	>5	nd	0.63	nd	nd	nd	0.68	>5
Anti-influenza (µg/ml)	>1	nd	>1	1.1	nd	nd	<1	nd	>1	nd	nd	nd	nd	nd	nd	>1
B cell population																
B lymphocyte ($CD19^+$; per µl × 10^{-3})	1.40	0.30	0.39–1.40	0.50	0.32	0.24	0.36	0.24	0.27–0.86	0.20	0.25	0.21	nd	nd	0.10	0.11–0.57
$\delta^+CD27^+/CD19+$ nonswitched (%)	nd	0.5	3–4	3.3	1.8	2.5	0.9	3.3	7–15	nd	2.9	1	nd	nd	nd	7–25
$IgM^{hi}CD38^+/CD19^+$ transitional (%)	nd	2	2–8	4	2.1	9.5	13	3	4–8	nd	14	13	nd	nd	nd	6–11
$CD27^+/CD19^+$ memory (%)	2	2	>10	5	4	5	2	5	>10	7	3	2	nd	nd	nd	>10
$\delta^-CD27^+/CD19^+$ switched (%)	1.9	0.7	4–10	2.2	2.2	2.4	0.6	0.1	4–18	nd	0.8	0.1	nd	nd	nd	10–28
Lymphocytes (per µl × 10^{-3})	4.9	2.1	2.3–5.4	3.1	2.3	2.6	2.4	1.6	1.9–3.7	1.7	1.8	2	nd	nd	1.1	1.4–3.3
T cell population (%)																
$CD3^+$	75	80	56–75	75	71	84	81	69	60–75	72	75	86	nd	nd	84	56–84
$CD4^+$	48	39	28–47	45	30	40	49	42	31–47	41	38	61	49	25	44	31–52
$CD8^+$	24	37	16–30	26	37	39	24	25	18–35	27	31	21	40	49	35	18–35
$CD31^+CD45RA^+/CD4^+$ naive	nd	35	50–85	60	nd	44	47	37	42–74	39	23	30	nd	nd	12	43–55
T cell proliferation (cpm × 10^{-3})																
Upon PHA stimulation	30	2.5	>50	47	30	41	27	52	>50	133	38	64	111	119	33	>50
Upon tetanus toxoid stimulation	6	9	>10	17	8	7	10	1	>10	1	10	nd	nd	nd	6	>10
Natural killer cells																
$CD56^+CD16^+$ (per µl × 10^{-3})	0.21	0.21	0.13–0.72	0.34	0.25	0.15	0.08	0.18	0.10–0.48	0.19	0.16	0.08	nd	nd	0.01	0.07–0.48

nd, not done. Patient VI-9 lacks a detailed immunological analysis, and thus was not included in the table.
[a]No increase after immunization.

Table S2. *POLE1* primers

Name	Primer
Genomic	
Exon 1F	5′-CAGAGGTGGTAGCCAACG-3′
Exon 1R	5′-GTTTCCCCGCAAAGAAGC-3′
Exon 2+3F	5′-CAGAGCAAGACTCCGTCTCA-3′
Exon 2+3R	5′-GGGTTTTAGCTTGTCGCAGT-3′
Exon 4-6F	5′-AGGAGCAAGCAATGTGTTTT-3′
Exon 4-6R	5′-CAGAGCCAGCCATTAAGGTA-3′
Exon 7+8F	5′-TTTCCTCGGTTTGAACTCTG-3′
Exon 7+8R	5′-TTTCACCATGTCCCTTCACT-3′
Exon 9+10F	5′-GGCCTAATGGGGAGTTTAGA-3′
Exon 9+10R	5′-ACGGTCATACCCTGAGAACA-3′
Exon 11+12F	5′-ACTTTGGGAGAGGAATTTGG-3′
Exon 11+12R	5′-CCATACTCTTGGGTGACCTG-3′
Exon 13+14F	5′-TCATCCTGGCTTCTGTTCTC-3′
Exon 13+14R	5′-CTCCTGTGGTTTCTTCCTCA-3′
Exon 15+16F	5′-ACCACGAGGTTTTCTCTCCT-3′
Exon 15+16R	5′-ACACAGACTGGCTCTTCCTG-3′
Exon 17-19F	5′-TGTCTAAAAGGGGTTGGTGA-3′
Exon 17-19R	5′-GAGCAGGAGCCACATCTTTA-3′
Exon 20F	5′-CTGAGCTGTTGCTCCTTTGT-3′
Exon 20R	5′-CCTTTAGGGTCCTTCTGAGG-3′
Exon 21F	5′-ATCTGTGAGGTGCTCCATGT-3′
Exon 21R	5′-ACTCTGCAAGTCCCTGAGTG-3′
Exon 22+23F	5′-GCATCTCTCCTTTCCTCCTC-3′
Exon 22+23R	5′-CTGGGAGCTATGCTGAAAGA-3′
Exon 24+25F	5′-TTGTCAATCCATCCACTCCT-3′
Exon 24+25R	5′-CCTTCTTGCTTCATCCTTCA-3′
Exon 26F	5′-GCCTAGAGGAGCACAGTCCA-3′
Exon 26R	5′-CACTTATGCCATCCCCAAAC-3′
Exon 27-29F	5′-CACGTCCTTATTTCCATGTTCA-3′
Exon 27-29R	5′-AAGCCTGGAGTCCTGTGTGT-3′
Exon 30+31F	5′-AGCCCGAGATCCTGAGATTT-3′
Exon 30+31R	5′-CTCCCCTTGGATCAAGGTCT-3′
Exon 32F	5′-GCAAGCTACCTGGAAGGTG-3′
Exon 32R	5′-GGAGGCCAGGCTAGATCAT-3′
Exon 33-35F	5′-GCCCAGTGACGAGATCAT-3′
Exon 33-35R	5′-TGTCCTCCTTCCCCAAAC-3′
Exon 36-38F	5′-TGCTTGTCGTGATTGAATTG-3′
Exon 36-38R	5′-TCACAGAATGGCAGAAACAC-3′
Exon 39F	5′-GTCTGTTGCTGGTTCTGGAG-3′
Exon 39R	5′-GGACCCTGTCTTAGACCTTAGC-3′
Exon 40F	5′-ACCTGTGCCCATTTCAGTT-3′
Exon 40R	5′-TTGGATTGTTATGCTCCACA-3′
Exon 41F	5′-AGGCTTTTCTGCTTCAGGAC-3′
Exon 41R	5′-CTTTTCACGCTGCTCAGATT-3′
Exon 42F	5′-GAGGGATGATGTGGTCTGAA-3′
Exon 42R	5′-GGTGCAGTGTCTGCTGCT-3′
Exon 43F	5′-GATGGACCCAGGTTGGAG-3′
Exon 43R	5′-CTGTCTCCCTTCTTGCGATA-3′

Table S2. *POLE1* primers *(Continued)*

Name	Primer
Exon 44+45F	5′-CCTCATCTGTGAGGCAATCT-3′
Exon 44+45R	5′-ATTACAGCCTCACCTTGCAC-3′
Exon 46+47F	5′-GCCTAAGGTCCAGAGGGTTC-3′
Exon 46+47R	5′-TCAGGACCTGCACACACC-3′
Exon 48+49F	5′-GCTCGAGGCTTACTGATGG-3′
Exon 48+49R	5′-CAGTGGTCTGGTCACTGGA-3′
RT–PCR	
cDNA Exons 32-37F	5′-TTCTACGTGAACCAGCGAGT-3′
cDNA Exons 32-37R	5′-TGGACTGAACAGCGATGAG-3′
Quantitative RT–PCR	
cDNA Exons 28-29F	5′-CGCCATCCAGAAGATCATCA-3′
cDNA Exons 28-29R	5′-CACGTGGCACTGGGTTCTTT-3′
probe cDNA Exons 28-29	5′-ATCCCTGCGGCCCTGCAGC-3′
cDNA Exons 33-34F	5′-GAGGGCGTATATGAGACTCAGGTT-3′
cDNA Exons 33-34R	5′-ACCAGCTGTTTATTGACCACACA-3′
probe cDNA Exons 33-34	5′-TTCCGGGCCCTGGTGCACC-3′
shRNA	
POLE-shRNA F	5′-GCAGTGGATTACTACTTTATT-3′
POLE-shRNA R	5′-AATAAAGTAGTAATCCACTGC-3′
Scramble-shRNA F	5′-GACTCGGGTAGTTTATGGA-3′
Scramble-shRNA R	5′-TCCATAAACTACCCGAGTC-3′