

SUPPLEMENTAL MATERIAL

Badran et al., <https://doi.org/10.1084/jem.20160724>

Table S1. Hemogram, inflammatory markers, and autoantibodies in the proband at 5 yr of age

Clinical status	Mildly symptomatic	Disease flare	Remission	Reference range ^{a,b}
Therapy, dose, (mg/kg)	Prednisolone, 0.3	Prednisolone, 0.19	Infliximab, 5	
Hemogram				
Hemoglobin (g/dl)	11.5	10.9	11.1	11.1–12.9
WBCs (10 ³ cells/ μ l)	8.35	22.3	9.5	6.0–10.8
Neutrophils (10 ³ cells/ μ l)	5.97	18.4	5.5	2.3–6.4
Lymphocytes (10 ³ cells/ μ l)	1.98	3.2	3.4	1.5–3.8
Monocytes (10 ³ cells/ μ l)	0.43	0.67	0.48	0.5–1.1
Platelets (10 ³ cells/ μ l)	270	260	238	190–365
Inflammatory markers				
CRP (mg/dl)	1.80	13.84	0.09	<0.50
ESR (mm/h)	28	37	12	0–20
Autoantibodies				
Antinuclear antibody	Negative	-	-	Negative
Ribonucleic protein IgG (AU/ml)	0	-	-	<40
Scleroderma-70 IgG (AU/ml)	0	-	-	<40
Smith, ENA IgG (AU/ml)	0	-	-	<40
SSA/RO, ENA IgG (AU/ml)	39	-	-	<40
SSB/LA, ENA IgG (mg/dL)	0	-	-	<40
Lymphocyte subsets				
CD3 ⁺ (10 ³ cells/ μ l)			3.6	1.4–3.7
CD3 ⁺ CD4 ⁺ (10 ³ cells/ μ l)			2.6	0.7–2.2
CD3 ⁺ CD8 ⁺ (10 ³ cells/ μ l)			0.82	0.49–1.3
CD19 ⁺ (10 ³ cells/ μ l)			0.67	0.39–1.4
CD16 ⁺ /CD56 ⁺ (10 ³ cells/ μ l)			0.30	0.13–0.72
Immunoglobulins				
IgG (mg/dl)			628	441–1,135
IgA (mg/dl)			133	22–159
IgM (mg/dl)			102	47–200
IgE (mg/dl)			2	0–200
Vaccine titers				
Pneumococcal IgG (number of positive subtypes)			7/14	\geq 7/14
Tetanus IgG (IU/ml)			2.25	>0.15
Polysaccharide ribose phosphate (ng/ml)			9,000	>1,000
Lymphocyte proliferation (cpm)				
PHA			338,981	104,415–319,780
Anti-CD3			120,069	78,423–205,485
Concanavalin A			214,659	74,586–194,337
Mitogen background			707	321–2,510
<i>Candida albicans</i>			30,053	20,561–189,605
Tetanus toxoid			46,584	14,938–96,819
Antigen background			4,157	724–7,752

^aThe reference range for the hemogram and inflammatory markers established by Boston Children's Hospital, where the test was performed.

^bThe reference range for autoantibodies established by ARUP Laboratories, University of Utah, where the test was performed.

Table S2. Heterozygous mutations shared between P1 and P2 identified by WES and not found in the Exome Aggregation Consortium database, the dbSNP database, 1000 Genomes database, or in our in-house collection of 80 exomes

Chr	Position	Strand	Reference	P1 and P2	Gene
chr01	47182041	-	C	T	<i>EFCAB14</i>
chr01	55058271	+	A	C	<i>ACOT11</i>
chr01	183212295	+	T	G	<i>LAMC2</i>
chr03	149498079	-	G	A	<i>ANKUB1</i>
chr06	137525482	-	T	C	<i>IFNGR1</i>
chr11	65427136	-	G	A	<i>RELA</i>
chr19	7688733	-	G	A	<i>XAB2</i>

Nucleotides from the coding DNA strand are listed. The patients' mutation in *RELA* is bolded.

Table S3. Characterization of the cellular infiltrate in the skin of WT and *Rela*^{+/-} mice after s.c. injection of TNF-α

Cell type	Staining marker	Number of cells (10 ³ /cm ²)			
		Saline		TNF	
		WT	<i>Rela</i> ^{+/-}	WT	<i>Rela</i> ^{+/-}
Dermis					
Neutrophils	CD11b ⁺ GR1 ^{high}	6.87 ± 3.30	2.70 ± 0.07	2.94 ± 0.62	29.69 ± 7.70*
Macrophages	CD11b ⁺ F4/80 ⁺	5.14 ± 3.02	2.72 ± 0.50	4.69 ± 1.74	11.72 ± 1.72*
DCs	CD11c ⁺ MHCII ^{high}	9.10 ± 2.69	6.48 ± 0.93	2.82 ± 0.29	2.24 ± 0.53
CD4 ⁺ T cells	CD3 ⁺ CD4 ⁺	0.17 ± 0.03	0.13 ± 0.03	0.10 ± 0.01	0.11 ± 0.04
CD8 ⁺ T cells	CD3 ⁺ CD8 ⁺	0.46 ± 0.16	2.07 ± 1.55	0.38 ± 0.20	0.48 ± 0.15
Hypodermis					
Neutrophils	CD11b ⁺ GR1 ^{high}	12.99 ± 9.17	4.06 ± 1.10	4.97 ± 1.69	1,039.00 ± 634.12
Macrophages	CD11b ⁺ F4/80 ⁺	7.53 ± 2.75	5.43 ± 1.36	8.44 ± 0.89	188.36 ± 10.21****
DCs	CD11c ⁺ MHCII ^{high}	14.74 ± 7.78	6.14 ± 0.97	1.86 ± 0.36	18.22 ± 2.00**
CD4 ⁺ T cells	CD3 ⁺ CD4 ⁺	0.04 ± 0.01	0.03 ± 0.02	0.02 ± 0.01	0.19 ± 0.04*
CD8 ⁺ T cells	CD3 ⁺ CD8 ⁺	1.06 ± 0.07	0.67 ± 0.61	0.26 ± 0.05	2.28 ± 0.61*

Data are means ± SEM. Asterisks indicate a significant difference between *Rela*^{+/-} and WT mice treated with s.c. TNF. *, P < 0.05; **, P < 0.01, ****, P < 0.0001, Student's t test. Data are pooled from two independent experiments, each having three 6–8-wk-old mice of each genotype.