Supplemental Tables

Supplemental Table 1. Haematological characteristics of the two patients in our study

	Patient 1				Patient 2			
	At birth	Infancy	Early toddler	Toddler	At birth	Infancy	Early toddler	Toddler
Height, cm, SD	32, −2.9	52, −6.0	67, −4.0	74, −3.1	43, −1.9	54, −2.5	66, −3.3	71, −2.6
Weight, g	726	3550	8210	11,750	2124	4465	7315	7945
BMI, SD	7, −5.2	13, −3.1	18, 1.9	21, 4.0	11, −1.0	15, −0.9	17, 0.5	16, 0.1
Laboratory findings								
Hgb, g/L	43	77*	64*	84*	54	70*	79*	67*
RBC, 10^{12}/L	0.95	2.48*	1.89*	2.70*	1.71	2.49*	2.09*	2.05*
MCV, fL	123.0	97.5	102.8	95.3	116.8	84.7	115.5	101.7
MCH, pg	45.1	31.2	33.8	31.1	32.4	27.9	37.6	32.4
MCHC, g/L	365	320	328	326	278	330	326	319
Reticulocytes, %, 10^9/L	16.6, 158	2.1, 51	2.9, 55	2.0, 53	1.7, 30	1.4, 27	0.4, 9	9.9, 203
Platelets, 10^9/L	396	929	465	418	306	670	474	283
WBC, 10^9/L	2.3	14.7	4.1	5.9	12.8	4.81	7.16	4.67
Neutrophils, 10^9/L	0.7	9.6	2.2	5.2	6.6	2.0	2.9	2.4
Lymphocytes, 10^9/L	1.3	4.6	1.7	0.5	3.6	2.2	3.4	2.0
Eosinophils, 10^9/L	0.0	0.1	0.1	0.0	0.2	0.3	0.4	0.3
Monocytes, 10^9/L	0.3	0.4	0.1	0.2	1.2	0.3	0.4	0.2
LDH (U/L)	565	293	263	352	2205	331	449	340
Total bilirubin (mg/L)	28.0	2.7	3.5	3.6	26.0	5.9	9.7	7.9
Bone marrow findings								
Cellularity		Normo-	Hyper-	Hyper-		Normo-	Hyper-	Hyper-
M:E ratio		1.5	0.5	2.0		1.5	0.9	1.5
Blasts, %		3.6	1.0	7.5		1.6	8.0	4.2
Dysplastic changes								
Erythroid cells		+	+	+		+	+	+
Myeloid cells		–	+	+		–	+	+
Megakaryocytes		–	+	+		–	+	+
G-banding	46,XX				46,XX			
Chromosome breakage testing	NA				Negative			

BMI, body mass index; Hgb, haemoglobin; LDH, lactate dehydrogenase; MCH, mean corpuscular haemoglobin; MCHC, mean corpuscular haemoglobin concentration; MCV, mean corpuscular volume; M:E, myeloid:erythroid; NA, not available; RBC, red blood cells; SD, standard deviation; WBC, white blood cells.

* Hgb levels and red blood cell counts were modified due to repetitive transfusion of red blood cells.

+ Dysplastic changes were detected. – Dysplastic changes were not detected.

Supplemental Table 2. Immunological characteristics of the two patients in our study

	Patient 1 (Infancy)	Reference interval	Patient 2 (Toddler)	Reference interval
Absolute lymphocyte count (cells/μL)	1200	(3400-9000)*	2400	(3600-8900)*
Lymphocyte subsets				
$CD3^+$ T cells (/μL, %)	690, 58	(1900-5900, 49-76)*	1169, 49	(2100-6200, 53-75)*
$CD4^+$ T cells (/μL, %)	576, 48	(1400-4300, 31-56)*	698, 29	(1300-3400, 32-51)*
$CD8^+$ T cells (/μL, %)	90, 7	(500-1700, 12-24)*	217, 9	(620-2000, 14-30)*
$CD19^+$ B cells (/μL, %)	341, 28	(610-2600, 14-37)*	240, 10	(720-2600, 16-35)*
$CD16^+$ $CD56^+$ NK cells (/μL, %)	110, 9	(160-950, 3-15)*	941, 39	(180-920, 3-15)*
Immunoglobulin profile				
IgG level (mg/dL)	377	(290-950)†	774	(470-1210)†
IgA level (mg/dL)	60	(8-50)†	39	(14-98)†
IgM level (mg/dL)	46	(46-176)†	65	(81-314)†

* Shearer WT, Rosenblatt HM, Gelman RS, et al. Lymphocyte subsets in healthy children from birth through 18 years of age: the Pediatric AIDS Clinical Trials Group P1009 study. J Allergy Clin Immunol.

† These reference ranges are derived from reference intervals of clinical tests in Japanese children (Tanaka T, Yamashita A, Ichihara K. 2008. Reference intervals of clinical tests in children determined by a latent reference value extraction method. J Jpn Pediatr Soc 112: 1117-1132 [in Japanese]).

CD, cluster of differentiation: NK, natural killer; Ig, immunoglobulin.

Supplemental Table 3. Clinical features of the two patients in our study compared to those with FILS and IMAGE-I syndromes

	Patient 1	Patient 2
FILS syndrome-compatible		
Facial dysmorphism	+	+
Immunodeficiency	–	–
Livedo	–	–
Short stature	+	+
IMAGE-I syndrome-compatible		
Intrauterine growth retardation	+	+
Metaphyseal dysplasia	+	–
Adrenal hypoplasia congenita	–	–
Genital anomalies	–	–
Immunodeficiency	–	–
Other features of our patients		
Congenital anaemia	+	+
Myelodysplastic syndrome	+	+
Inflammatory bowel disease	+	–
Microcephaly	+	–
Pulmonary fibrosis	–	–
Nail dystrophy	–	–
Teeth abnormality	–	–
Hypogammaglobulinemia	–	–
Splenomegaly	–	–

FILS, facial dysmorphism, immunodeficiency, livedo, and short stature; IMAGE-I, intrauterine growth restriction, metaphyseal dysplasia, adrenal hypoplasia congenita, genital anomalies and immunodeficiency