MATERIAL SUPPLEMENTARY

Table 1. Laboratory features of the patient

	Nov. 2011	June 2013	Jan. 2014	July 2015	Sep. 2018
White blood cell count/mm3	19900	9340	11000	4150	6630
Absolute neutrophil count/mm3	11600	4420	4360	2290	3410
Hemoglobin (g/dl)	10,9	10,9	10,9	109	11,2
Platelets /mm3	1.080.000	578000	732000	501000	344000
C-reactive protein (mg/dl)	2,5	<0,33	0,8	1	0,3
IgA (mg/dl)	<5	6,5	12,3	38,9	41,6
IgG (mg/dl)	1030	529	520	915 (under IVIG replacement)	1110
IgM (mg/dl)	223	75	72,8	55,7	72,9
CD3+ T cells	62%	71%		63%	
CD3+CD4+ Thelper cells	38%	41%		38%	
CD3+CD8+ T cells	25%	25%		22%	
CD3-CD16/56+ natural killer cells	5%	6%		5%	
CD19+ B cells	25%	21%		25%	
CD19+IgM-CD27+ memory B cells		38%		54%	

Table 2. Summary of clinical signs of THES patients

Summary of clinical signs of THES patients reported before		Clinical signs of the index patient
Constant findings		
1. Intractable diarrhea of infancy	*chronic intractable diarrhea beginning at newborn period, need for a parenteral nutrition*	Mild, intermittent diarrhea following respiratory tract infections
2. Facial dysmorphism	*hypertelorism, broad flat nasal bridge and prominent forehead*	Flat broad nose, prominent forehead, large mouth and large ears
3. Hair abnormalities	*coarse, sparse, easily removable, brittle, trichorrhexis nodosa*	Coarse, brittle hair with trichorrhexis nodosa
Very frequent findings		
4. Immunodeficiency	*defective specific antibody responses despite normal serum immunoglobulin levels, hypogammaglobulinemia, low $CD27^+$ memory B cells, low lymphocyte counts, T cell proliferation defects.*	Low IgG levels
5. Intauterin growth restriction	*IUGR or small for gestational age*	-
Frequent findings		
6. Skin abnormalities	*hypopigmentation, café au lait spots, xerosis*	Xerosis, pyoderma gangrenosum like skin eruptions, oral aphtous lesions
7. Liver disease	*hepatomegaly, fibrosis, hemosiderosis*	-
Rare findings		
8. Congenital cardiac defects	*ventricular septal defect, atrial septal defect, peripheral pulmonary stenosis, tetralogy of Fallot*	tricuspid and pulmonic valve regurgitation
9. Platelet anomaly	*enlarged platelets, thrombocytosis*	Intermittent thrombocytosis, normal MPV levels
10. Mild developmental delay		-
11. Tooth abnormality	*Not reported*	Peg-teeth anomaly, short roots

Figure 1. (a) Pyoderma gangrenosum like skin lesions, (b) rough and brittle hair, (c) short tooth roots with «peg teeth» anomaly and (d) light microscopic examination of scalp hair showing *trichorrhexis nodosa*

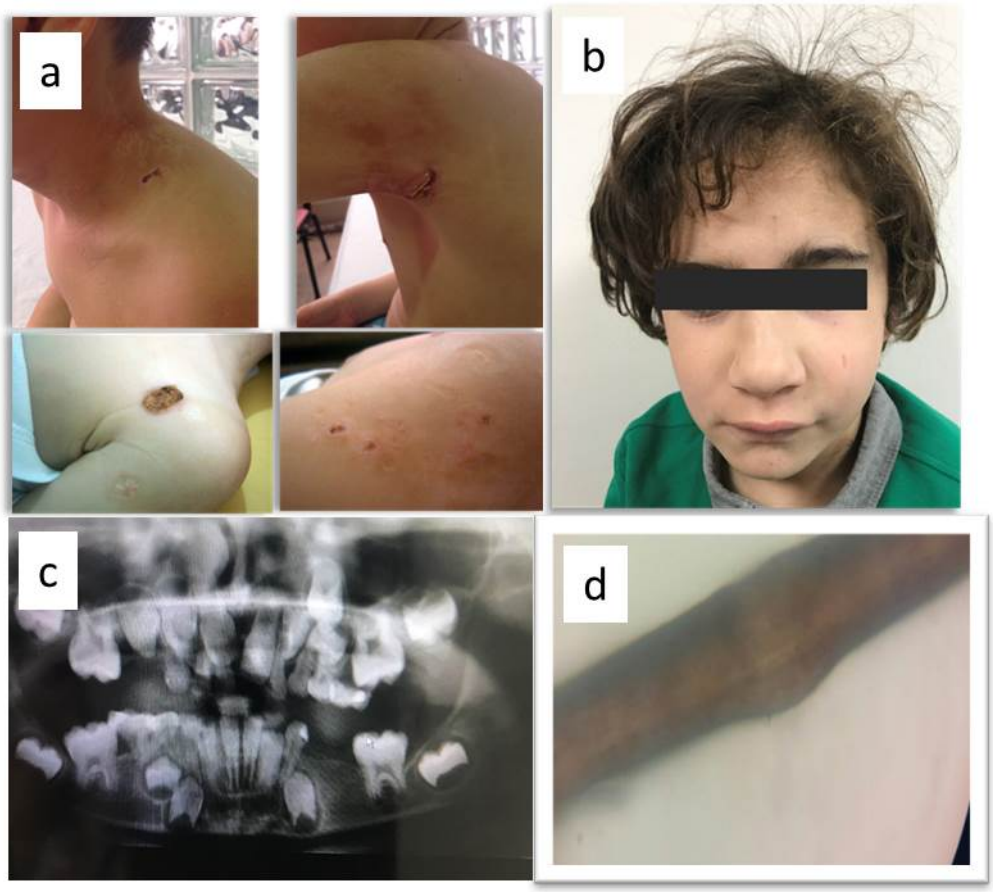

Figure 2. Sequencing analysis showing a homozygous mutation in exon 21 of *TTC37* gene (c.2210T>C, p.Val737Ala)

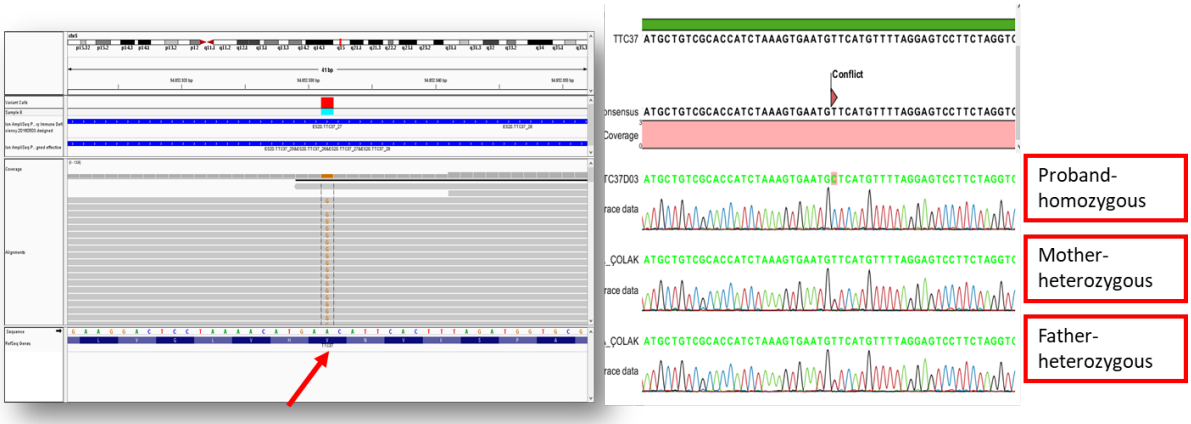

Figure 3. Growth chart of the patient