

Corrigendum

A human immunodeficiency caused by mutations in the *PIK3R1* gene

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The authors recently identified an error in the identification numbers assigned to patient samples that resulted in an incorrect clinical and immunological description of patient 4 (P4). The error did not affect the conclusions of the manuscript; however, the correct immunological profile is now included in the revised version of Table 1, which appears below.

Corrected text for the clinical description is provided below:

Results and Discussion, 1st paragraph:

All 4 patients had been suffering from recurrent respiratory bacterial tract infections from soon after birth, although none of them was hospitalized to treat infections (Table 1 and Supplemental Figure 1; supplemental material available online with this article; doi:10.1172/JCI75746DS1). They had no signs of allergy, autoimmunity, splenomegaly, or lymphadenopathy (with the exception of patient 1 [P1], whose tonsils had been removed in 2 separate operations). Although viral infections and opportunistic bacterial or fungal infections were not reported, P2 and P4, but not P1, presented with EBV viremia, as detected by PCR (P2, 1,500 copies per ml; P4, 540 copies per ml; reference values, <182, respectively). P2 also presented with CMV viremia (CMV, 9,300 copies per ml; reference values, <446). Lymphocyte counts were in the normal range, but the frequency of CD31⁺CD45RA⁺ naive CD4 and CCR7⁺CD45RA⁺ naive CD8 peripheral blood T cells was low in all patients (Table 1). An elevated proportion of CD8 T cells expressing the senescence-associated marker CD57 was observed in the 3 infant patients (P1, P2, and P4). P1 displayed a memory B cell deficiency. All patients had elevated frequency of transitional B cells (Table 1). Impaired B cell function was suggested by the serum Ig profile: undetectable IgA levels in all patients; very low IgG levels in P1 and P2 (but elevated levels in P3); and elevated IgM levels in all 4 patients. All the patients but P3 were receiving Ig replacement therapy. The immunological phenotype of these patients was somewhat heterogeneous and reminiscent of that observed in APDS. The presence of dominant gain-of-function mutations of *PIK3CD* was ruled out for all 4 patients (data not shown). P2 presented with growth retardation (–2 SD for weight and –2.5 SD for height), without improvement after treatment with Ig replacement therapy. P4 also presented with slight growth retardation (–1.5 SD for weight and –2 SD for height).

The authors regret the errors.

Table 1. Clinical and immunologic features

Patient (family)	P1 (family 1)	P2 (family 2)	Control values (age matched) (P1, P2)	P4 (family 3)	Control values (age matched) (P4)	P3 (family 2)	Control values (age matched) (P3)
Gender	Female	Male		Male		Female	
Age at presentation (yr)	2	0.6		4		0.6	
Viral infection	No	Enteroviral enteritis; chronic CMV and EBV (asymptomatic viremia)		Chronic EBV (asymptomatic viremia)	No		
Lymphoproliferation	Enlarged tonsils	No				No	
Autoimmunity	No	No		No		No	
Allergy	No	No		No		No	
Respiratory features (infections)	URT and LRT infections; recurrent conjunctivitis	URT and LRT infections		URT and LRT infections		URT infections	
Ig replacement therapy	+	+		+		-	
Total IgG, g/l (age in yr)	<0.07 (2)	<0.07 (2)	3.35–8.96 ^A	3.68 (4)	3.35–8.96 ^A	22.08^B (35)	5.49–12.78
IgA, g/l (age in yr)	<0.06 (2)	<0.06 (2)	0.27–1.22 ^A	0.1 (4)	0.27–1.22 ^A	<0.06 (35)	0.41–3.44
IgM, g/l (age in yr)	3.67 (2)	2.89 (2)	0.58–1.53 ^A	4.68 (4)	0.58–1.53 ^A	2.26 (35)	0.5–2.09
Age at analysis (yr)	4	4	2–6	14	12–16	34	18–35
T cells/ μ l	1,512	3,432	1,400–3,700	1,900	1,000–2,200	1,394	807–1,844
CD4 ⁺ T cells/ μ l	432	780	700–2,200	779	530–1,300	833	460–1,232
CD8 ⁺ T cells/ μ l	576	2,457	490–1,300	741	330–920	476	187–844
Naive CD4 ⁺ T cells (%)	39	19	57–65	25	43–55	4	43–55
Naive CD8 ⁺ T cells (%)	21	8	52–68	3	52–68	4	52–68
CD3 ⁺ CD8 ⁺ CD57 ⁺ (%)	16	9	<5	26	4–20	16	4–20
B cells/ μ l	101	312	390–1,400	19	110–570	119	92–420
Transitional B cells (%)	26	40	2–8	20	2–8	39	2–11
Memory B cells (%)	8	11	>10	18	>10	72	>10
Class-switched memory B cells (%)	25	29	>10	93	>10	23	>10
NK cells/ μ l	108	156	130–720	266	70–480	187	89–362

Values outside the normal range are given in bold. ^AValues reported are identical for control values (P1, P2) and control values (P4) because the age that measurements were taken is within the same range. ^BIgG1 = 6.6 (>4); IgG2 = 6.7 (>0.6); IgG3 = 0.78 (>0.17); IgG4 = 0.02 (0–2.10). The concentration of total IgG determined by the addition of IgG subclass ELISAs or total IgG determined by nephelometry can differ due to methodical variations. URT, upper respiratory tract; LRT, lower respiratory tract; ND, not determined; naive CD4⁺ T cells, CD31⁺CD45RA⁺/CD4⁺; naive CD8⁺ T cells, CCR7⁺CD45RA⁺/CD8⁺; transitional B cells, CD21⁺CD24⁺/CD19⁺; memory B cells, CD27⁺/CD19⁺; class-switched memory B cells, IgM⁺IgD⁺/CD27⁺CD19⁺; NK cells: CD16⁺CD56⁺.