

Table E1

Laboratory Evaluation	Reference Ranges	2014	2015	2016
WBC (K/mcL)	4.5-13.5K/mcL	2	4	2.4
Hgb (gm/dL)	12-16 gm/dL	11	12	13.9
PLT (K/mcL)	135-466K/mcL	92	178	91
Neutrophils	40-62%	15	40	31
Lymphocytes	15-45%	63	44	52
Monocytes	0-8%	16	11	9
Eosinophils	0-5%	4	4	8
Basophils	0-1%	1	1	0
ANC (mcL)	1.8-8K/mcL	0.3	1.6	0.74
ALC (mcL)	1.5-6.5K/mcL	1.2	1.76	1.25
Reticulocytes	0.5-1.5%	1.9	2.8	2
Direct Coombs	-	+	+	-
Anti-neutrophil Abs	-	+	+	+
Anti-PLT Abs	-	+	+	+
Anti-Class I/II Abs		-	+	+
Lymphocyte subsets				
CD3	52-78% 800-3500 cells/mcL	77 % 1309	77% 1243	78% 1092
CD4	25-48% 400-2100 cells/mcL	44% 694	43% 689	48% 663
CD8	9-35% 200-1200 cells/mcL	32% 545	29% 469	25% 349
CD19	(8-24%) 200-600 cells/mcL	21% 358	20% 324	20% 276
CD16/56	6-27% 70-1200	1% 17	2% 38	1% 19
CD4/CD8	1.0-3.6	1.3	1.5	1.9
FOXP3+Tregs	55-81%	68	58	61
CD4+25+127 low Tregs	4.2-9.9% of CD4+	4.4	4.4	4.4
TCRV11/VA24	>=0.01%	0.06		
CD4+CCR6+CD45RA-*	10.7-27% of CD4+	20.1	19.3	
CCR6+CD45RA-IL-17A+	1.2-4.6% of * cells	5.6	3.6	
ALPS Markers				
IL-10	<=7 pg/mL	5	1	6
IL-18	89-540 pg/mL	657	499	614
Vit B12	193-986 pg/mL	774	788	775
Fas-apoptosis cell loss	55-95%	81		97
TCR$\alpha\beta$-DNT cells*	> 2% of lymphocytes*	2.6	1.9	2.6
B220 TCR$\alpha\beta$-DNT cells*	> 60% of DNT cells*	19	22	32
CD3+CD25+HLA/DR ratio*	< 1*	1	1.1	1
CD27+ B cells*	< 15% of B cells*	8	8	9
IgG	600-1500 mg/dL	1150	On IVIG	On IVIG
IgM	60-263 mg/dL	54	71	166
IgA	68-378 mg/dL	200	213	234
IgE	2-214 mg/dL	3	-	3
B-cell Panel				
CD19+ B cells	8- 24 %	15	18	
CD19+CD27-CD21-IgM++	0.5- 2.8 %	6.7	5.7	
CD19+CD27-	61- 83 %	87	89	
CD19+CD27-IgM+IgD+	53- 79 %	83	85	
CD19+CD27-IgM+IgD+	80- 97 % of CD27-	95	95	
CD19+CD27-CD21-CD38-	0.3- 2.3 %	4.4	5	
CD19+CD27+	17- 39 %	13	11	
CD19+CD27+IgM+IgD+	8- 22 %	10	9	
CD19+CD27+IgM+IgD+	30-76 % of CD27+	79	78	
CD19+CD27+IgM-IgD-	2.3- 18.2 %	1.8	1.7	
CD19+CD27+IgM-IgD-	14-44 % of CD27+	14	15	

Table E1

Table E1. A. Immunological Evaluation at Referral and Follow-up Visits

Bold values represent abnormal values.

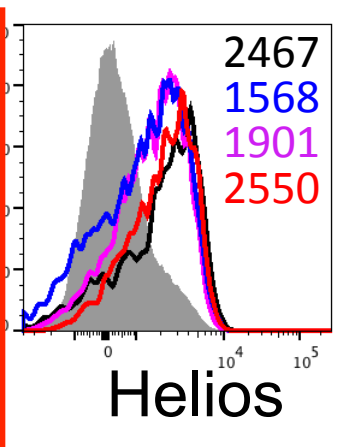
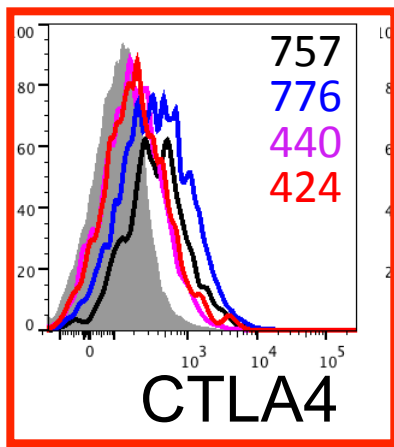
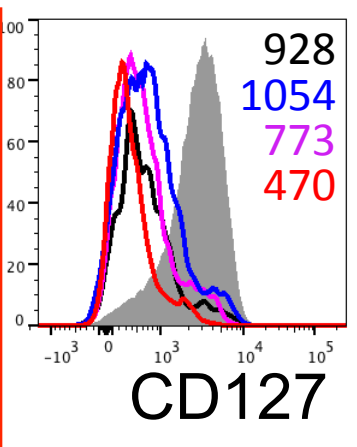
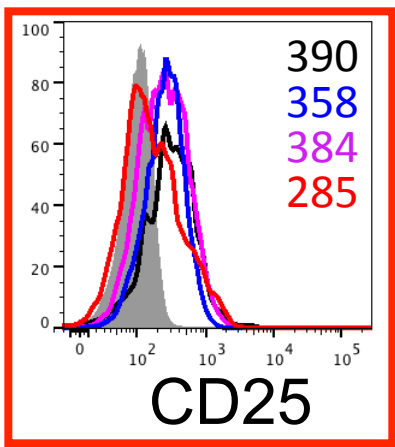
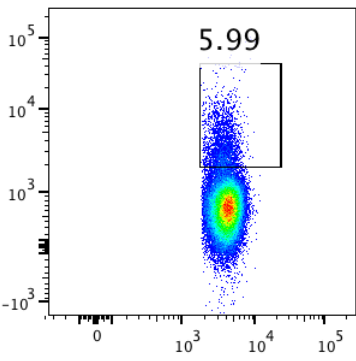
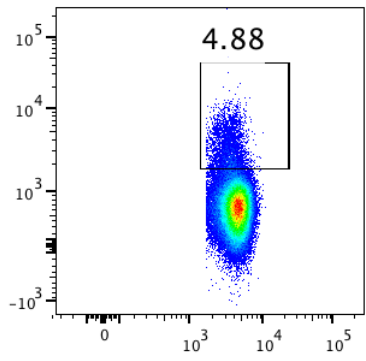
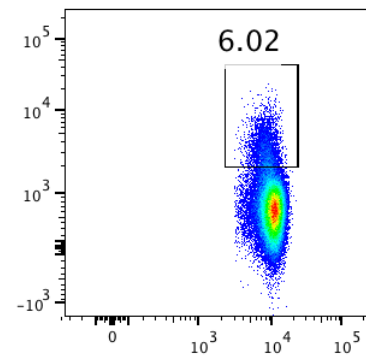
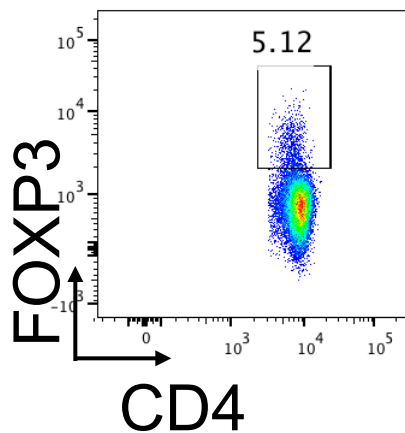
* Parameters specific for flow cytometry based ALPS panel. Reference values indicate positive ALPS score for each parameter.

HC

Father

Mother-mutant
asymptomatic

Patient



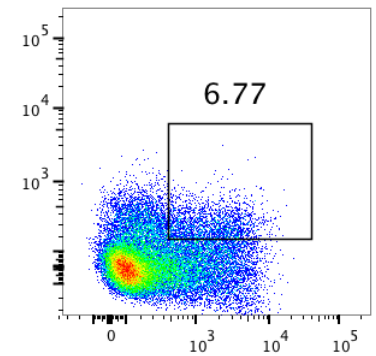
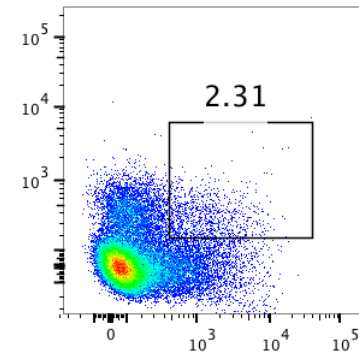
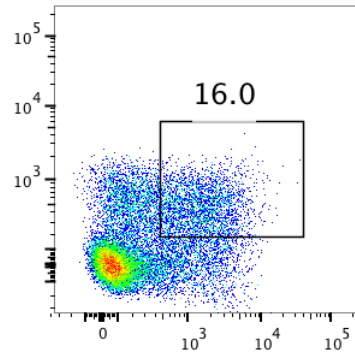
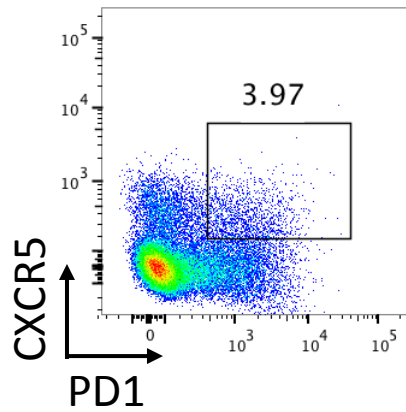
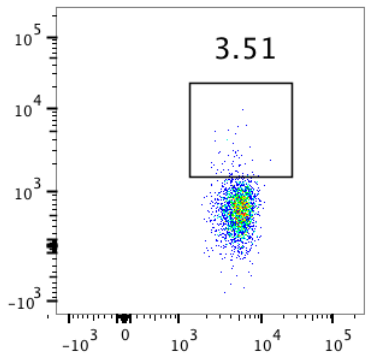
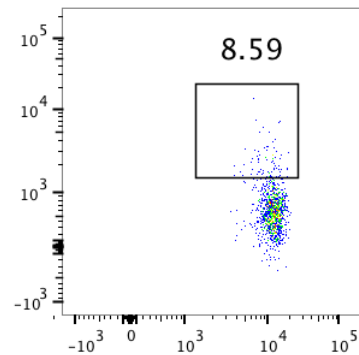
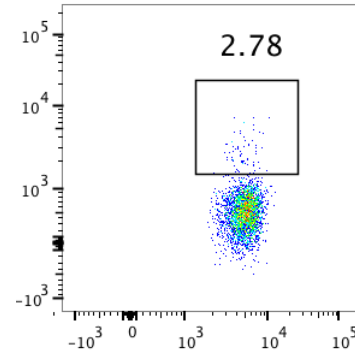
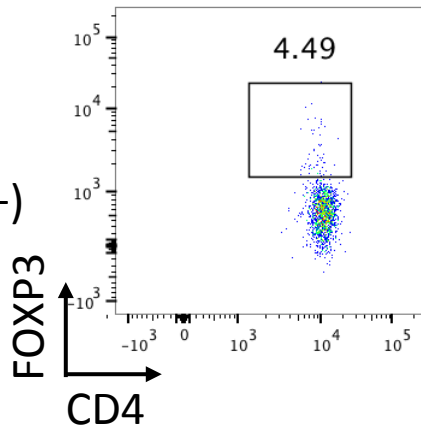
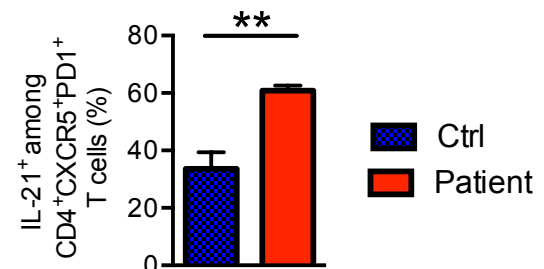
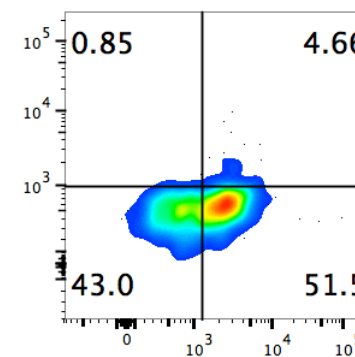
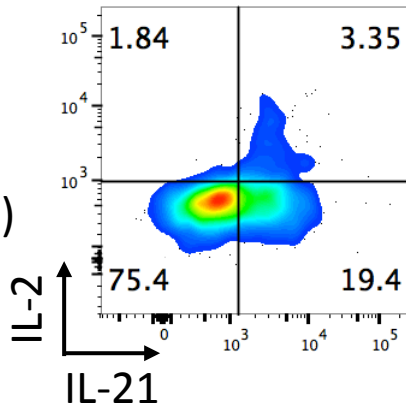
— T_{REG} HC
 — T_{REG} Father
 — T_{REG} Mother
 — T_{REG} Patient

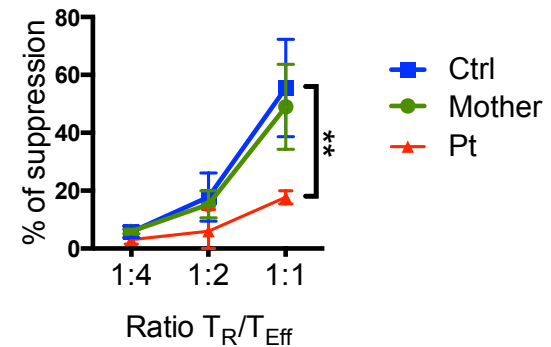
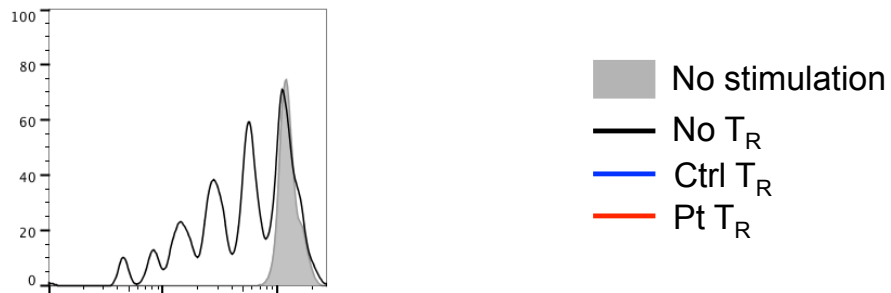
Ctrl

Patient

Father

Mother-carrier

Gated on
CD4⁺ T cellsGated on
T_{FH} cells
(CXCR5+PD1+)Gated on
T_{FH} cells
(CXCR5+PD1+)



Ratio T_R/T_{Eff}

