

Table S1: Patient Characteristics

Patient ID	Age (years)	RMRP Mutation	Recurrent or severe infections	Atopic Disease	GI Manifestations	Joint Symptoms	ALC (cells/ μ L)	CD3+ (cells/ μ L)	CD19+ (cells/ μ L)
CHH004	39	70A>G/70A>G	+	+	-	-	1520	770	170
CHH009	69	70A>G/70A>G	+	+	-	-	1310	960	90
CHH012	26	70A>G/262G>T	-	+	-	-	940	810	40
CHH016	63	70A>G/70A>G	+	+	-	+	2010	1550	190
CHH017	51	70A>G/70A>G	-	+	+ Gastritis	+	1450	950	160
CHH019	28	70A>G/70A>G	-	+	+ Lactose intolerance, digestive difficulty	-	1700	1540	230
CHH020	23	70A>G/70A>G	+	+	+ Bloody diarrhea of unknown etiology	-	940	450	120
CHH023	59	70A>G/70A>G	+	+	+ Constipation	-	1010	630	30
CHH029	63	70A>G/70A>G	+	+	+ Oral aphthous ulcers, abdominal pain	-	1190	1110	140
CHH033	65	70A>G/262G>T	+	+	-	-	2010	1360	120
CHH035	45	70A>G/70A>G	+	-	-	+	1500	1250	110
CHH037	23	70A>G/70A>G	+	-	+ Lactose intolerance	-	640	500	100
CHH043	58	70A>G/70A>G	+	-	-	-	1250	590	90
CHH079	37	70A>G/70A>G	-	-	+ Occasional loose stools of unknown etiology	-	2220	1910	270
CHH100	44	70A>G/70A>G	+	+	+ Diarrhea	-	1180	630	130
CHH130	32	70A>G/70A>G	+	+	+ Intestinal discomfort with certain foods	-	1160	NT	NT

NT = not tested