

	Patients	Gene	Comments	cDNA	Protein	Variant Classification
A (CVID)	Pt.1	CR2	Complement C3d Receptor 2	c.914G>T	p.Cys305Phe	VUS
	Pt.2	CR2	Complement C3d Receptor 2	c.826delT (he)	p.Cys276fs	Pathogenic
	Pt.2	PRF1	Perforin 1	c.272C>T (he)	p.Ala91Val	Likely Pathogenic
	Pt.2	TCF3	Transcription Factor 3	c.1331G>A (he)	p.Gly444Glu	VUS
	Pt.2	TNFRSF13B	Tumor Necrosis Factor Receptor Superfamily Member 13B	c.431C>G (he)	p.Ser444Ter	Pathogenic
	Pt.2	TNFRSF13B	Tumor Necrosis Factor Receptor Superfamily Member 13B	c.577T>C (he)	p.Cys193arg	VUS
	Pt.3	TNFRSF13B	Tumor Necrosis Factor Receptor Superfamily Member 13B	c.260T>A (he)	p.Ile87Asn	Likely Pathogenic
	Pt.4	AICDA	Activation Induced Cytidine Deaminase	c.361G>C (he)	p.Ala121Pro	VUS
	Pt.5	CECR1	Cat eye syndrome chromosome region, candidate 1	c.927G>A (he)	p.Met309Ile	VUS
	Pt.6	LRBA	Lipopolysaccharide-responsive beige-like anchor protein	c.8201T>G (he)	p.Leu2734Trp	VUS
B (ALPS)	Pt.7	TNFRSF6	Tumor Necrosis Factor Receptor Superfamily Member 6	c.568G>A (he)	p.Val190Met	Likely pathogenic
	Pt.8	TNFRSF6	Tumor Necrosis Factor Receptor Superfamily Member 6	c.856G>T (he)	p.Gly268Ter	Pathogenic
	Pt.9	TNFRSF6	Tumor Necrosis Factor Receptor Superfamily Member 6	c.580G>A (he) c.673T>A (he)	p.Glu194Lys p.Ser225Thr	Pathogenic VUS

Table 1 IEI-related molecular analysis through a specific IEI targeted gene panel

Abbreviations: ALPS, Autoimmune lymphoproliferative syndrome; CVID, Common variable immune deficiency; VUS, Variants of uncertain significance.