

Table 3 Pseudomyopathy Antibody Deficiencies	Hoffmann syndrome/TOPGB deficiency	TOPGB	AD	129431	P=not attempted	Immunoglobulin, supportive care for immunodeficiency, not due to IgA deficiency	- antidiarrheal	PMD 31402095
Table 3 Pseudomyopathy Antibody Deficiencies	Common variable immune deficiency with no gene defect specified (CVID)	Unknown	Variable		P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for diarrheal disease	PMD 30302700 PMD 32350032
Table 3 Pseudomyopathy Antibody Deficiencies	ARIHSE1 deficiency	ARIHSE1	AR	610450	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for diarrheal disease	PMD 30314465
Table 3 Pseudomyopathy Antibody Deficiencies	ATP6BP1 deficiency	ATP6BP1	XL	200131	P=not attempted	Immunoglobulin, pathogenic antibodies for hemophagocytosis	- antidiarrheal, surveillance for hemophagocytosis	PMD 27123100 PMD 32112104
Table 3 Pseudomyopathy Antibody Deficiencies	CD19 deficiency	CD19	AR	167455	P=not attempted	Immunoglobulin, pathogenic antibodies for hemophagocytosis	- antidiarrheal, surveillance for hemophagocytosis	PMD 30305520 PMD 3075250
Table 3 Pseudomyopathy Antibody Deficiencies	CD20 deficiency	CD20	AR	112110	P=not attempted	Immunoglobulin	- antidiarrheal	PMD 20305000
Table 3 Pseudomyopathy Antibody Deficiencies	CD21 deficiency	CD21	AR	120401	P=not attempted	Immunoglobulin, plasmazyme	- antidiarrheal, surveillance for diarrheal disease	PMD 22035000
Table 3 Pseudomyopathy Antibody Deficiencies	CD81 deficiency	CD81	AR	120843	P=not attempted	Immunoglobulin	- antidiarrheal, plasmazyme	PMD 30305000 PMD 20274010
Table 3 Pseudomyopathy Antibody Deficiencies	KAR205 hypofunction	KAR205	AD	602001	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for IgA deficiency	PMD 20081452
Table 3 Pseudomyopathy Antibody Deficiencies	MPBP2 deficiency	MPBP2	AD	611500	P=not attempted	Immunoglobulin, pathogenic antibodies for hemophagocytosis	- antidiarrheal, surveillance for hemophagocytosis	PMD 27012100
Table 3 Pseudomyopathy Antibody Deficiencies	Mannosidase/glycosidase deficiency (MGOB)	MGOB (GCS1)	AR	601100	P=not attempted	Immunoglobulin, supportive care for hemophagocytosis, dietary and recurrent fevers	- antidiarrheal, surveillance for hemophagocytosis	PMD 24711001
Table 3 Pseudomyopathy Antibody Deficiencies	MPBP2 deficiency	MPBP2	AD	610111	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for hemophagocytosis	PMD 30305000
Table 3 Pseudomyopathy Antibody Deficiencies	MPBP2 deficiency	MPBP2	AD	610112	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for hemophagocytosis	PMD 30305000
Table 3 Pseudomyopathy Antibody Deficiencies	PK3CD mutation (GOF) APD02	PK3CD GOF	AD	600830	P=not attempted	Immunoglobulin, immunosuppressive agents	- antidiarrheal, surveillance for IgA deficiency	PMD 21001110 PMD 21402010 PMD 21402010 PMD 21402010
Table 3 Pseudomyopathy Antibody Deficiencies	PK3CD mutation (LGP) APD02	PK3CD LGP	AD	600831	P=not attempted	Immunoglobulin, immunosuppressive agents	- antidiarrheal, surveillance for IgA deficiency	PMD 21001110 PMD 21402010 PMD 21402010 PMD 21402010
Table 3 Pseudomyopathy Antibody Deficiencies	PTEN Deficiency (LOF)	PTEN	AD	611720	P=not attempted	Immunoglobulin, immunosuppressive agents	- antidiarrheal, surveillance for IgA deficiency	PMD 21001110 PMD 21402010 PMD 21402010 PMD 21402010
Table 3 Pseudomyopathy Antibody Deficiencies	RAC11 deficiency	RAC11	AR	610500	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for IgA deficiency	PMD 20212001
Table 3 Pseudomyopathy Antibody Deficiencies	SEC61AT1 deficiency	SEC61AT1	AD	600911	P=not attempted	Immunoglobulin	- antidiarrheal	PMD 20202001
Table 3 Pseudomyopathy Antibody Deficiencies	SHOBP1 (CNIB) deficiency	SHOBP1	XL	200110	P=not attempted	Immunoglobulin	- antidiarrheal	PMD 20202001
Table 3 Pseudomyopathy Antibody Deficiencies	TACI deficiency	TACI	AD or AR	624001	P=not attempted	Immunoglobulin	- antidiarrheal, surveillance for IgA deficiency	PMD 20202001
Table 3 Pseudomyopathy Antibody Deficiencies	TAFF receptor deficiency	TNFRSF13C	AD	600443	P=not attempted	Immunoglobulin	- antidiarrheal	PMD 20202001
Table 3 Pseudomyopathy Antibody Deficiencies	TWEAK receptor deficiency	TNFRSF12	AD	600444	P=not attempted	Immunoglobulin	- antidiarrheal	PMD 20202001
Table 3 Pseudomyopathy Antibody Deficiencies	TRNT1 deficiency	TRNT1	AR	612001	P=not attempted	Immunoglobulin, biological therapy for autoinflammatory condition, complement	- antidiarrheal, fever, cutaneous and ocular dysfunctions	PMD 25193071
Table 3 Pseudomyopathy Antibody Deficiencies	AD deficiency	ACDA	AR	603051	P=not attempted	Immunoglobulin	- antidiarrheal, IgA deficiency	PMD 30305002
Table 3 Pseudomyopathy Antibody Deficiencies	AD deficiency	ACDA	AD	602012	P=not attempted	Immunoglobulin	- antidiarrheal, IgA deficiency	PMD 30305002
Table 3 Pseudomyopathy Antibody Deficiencies	IGK deficiency	IGK	AR	610500	P=not attempted	Immunoglobulin	- antidiarrheal, IgA deficiency	PMD 20212000
Table 3 Pseudomyopathy Antibody Deficiencies	IGKV deficiency	IGKV	AR	610501	P=not attempted	Immunoglobulin	- antidiarrheal, IgA deficiency	PMD 20212000
Table 3 Pseudomyopathy Antibody Deficiencies	related IgG subclass deficiency	Unknown	9		P=not attempted	Consider immunoglobulin replacement therapy	- IgG subclass may be considered	PMD 20081452 PMD 20080700
Table 3 Pseudomyopathy Antibody Deficiencies	IgG subclass deficiency with IgA deficiency	Unknown	9		P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20081452 PMD 20080700
Table 3 Pseudomyopathy Antibody Deficiencies	Selective IgG subclass deficiency	Unknown	9		P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20081452 PMD 20080700
Table 3 Pseudomyopathy Antibody Deficiencies	Specific antibody deficiency with normal Ig levels and normal B cells	Unknown	9		P=not attempted	Immunoglobulin replacement therapy	- IgG subclass may be considered	PMD 20081452 PMD 20080700
Table 3 Pseudomyopathy Antibody Deficiencies	Transient hypogammaglobulinemia of infancy	Unknown	9		P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20081452 PMD 20080700
Table 3 Pseudomyopathy Antibody Deficiencies	Selective IgM deficiency	Unknown	9		P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20081452
Table 3 Pseudomyopathy Antibody Deficiencies	CARD11 GOF	CARD11 AD GOF	607210	P=not attempted	Immunoglobulin, thymusectomy	- IgG subclass may be considered	PMD 23125000	
Table 3 Pseudomyopathy Antibody Deficiencies	Kappa chain deficiency	IGKC	AR	147400	P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20202000
Table 3 Pseudomyopathy Antibody Deficiencies	h chain mutations and deletions	IGK	AD	610500	P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20202000
Table 3 Pseudomyopathy Antibody Deficiencies	FAAP24 deficiency	FAAP24	AR	610884	P=not attempted	Immunoglobulin	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Perforin deficiency (PHLZ)	PRF1	AR	112001	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 27170309
Table 4 Diseases of Immune Dysregulation	Syntactic protein intolerance SLC7A1 deficiency	SLC7A7	AD	222700	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Transmembrane protein 140 (TMEM140) deficiency	TMEM140	AD	222701	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	TMEM192 (Mac-2 related protein) deficiency (HLR3)	TMEM192 HLR3	AD or AD	222702	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	UNC-303 (Monocyte-14-like) deficiency (HLR3)	UNC-303	AD	222703	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Hemophagocytosis-Puttak syndrome, type 2	APPB1	AD	600401	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Hemophagocytosis-Puttak syndrome, type 10	APPD11	AD	610500	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Chediak-Higashi syndrome	LYST	AR	600893	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	Glycine dehydrogenase, type 2	KABT2A	AR	603884	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	BACH1 deficiency	BACH2	AD	602041	P=not attempted	HLH therapies	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CTLA4 deficiency (ALPSV)	CTLA4	AD	122800	P=not attempted	Adolescent, avitilavimab	- IgG subclass may be considered	PMD 20 29729943
Table 4 Diseases of Immune Dysregulation	DefB deficiency	DEFB	AR	610004	P=not attempted	Adolescent, avitilavimab	- IgG subclass may be considered	PMD 31300374
Table 4 Diseases of Immune Dysregulation	FERMT1 deficiency (Klenerman syndrome)	FERMT1	AR	122800	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	PEX1, immune dysregulation, polyendocrinopathy, enteropathy X-linked	PEX1	AD	200200	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD5 deficiency	CD5	AD	147400	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD123 deficiency	CD123	AD	600400	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD163 deficiency	CD163	AD	600401	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD164 deficiency	CD164	AD	600402	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD166 deficiency	CD166	AD	600403	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD167 deficiency	CD167	AD	600404	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD168 deficiency	CD168	AD	600405	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD169 deficiency	CD169	AD	600406	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD170 deficiency	CD170	AD	600407	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD193 deficiency	CD193	AD	600408	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD194 deficiency	CD194	AD	600409	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD195 deficiency	CD195	AD	600410	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD196 deficiency	CD196	AD	600411	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD197 deficiency	CD197	AD	600412	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD198 deficiency	CD198	AD	600413	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD199 deficiency	CD199	AD	600414	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD200 deficiency	CD200	AD	600415	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD201 deficiency	CD201	AD	600416	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD202 deficiency	CD202	AD	600417	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD203 deficiency	CD203	AD	600418	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD204 deficiency	CD204	AD	600419	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD205 deficiency	CD205	AD	600420	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD206 deficiency	CD206	AD	600421	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD207 deficiency	CD207	AD	600422	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD208 deficiency	CD208	AD	600423	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD209 deficiency	CD209	AD	600424	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD210 deficiency	CD210	AD	600425	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD211 deficiency	CD211	AD	600426	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD212 deficiency	CD212	AD	600427	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD213 deficiency	CD213	AD	600428	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD214 deficiency	CD214	AD	600429	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD215 deficiency	CD215	AD	600430	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD216 deficiency	CD216	AD	600431	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD217 deficiency	CD217	AD	600432	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD218 deficiency	CD218	AD	600433	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD219 deficiency	CD219	AD	600434	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD220 deficiency	CD220	AD	600435	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD221 deficiency	CD221	AD	600436	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD222 deficiency	CD222	AD	600437	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD223 deficiency	CD223	AD	600438	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD224 deficiency	CD224	AD	600439	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD225 deficiency	CD225	AD	600440	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD226 deficiency	CD226	AD	600441	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD227 deficiency	CD227	AD	600442	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD228 deficiency	CD228	AD	600443	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD229 deficiency	CD229	AD	600444	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD230 deficiency	CD230	AD	600445	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD231 deficiency	CD231	AD	600446	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD232 deficiency	CD232	AD	600447	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD233 deficiency	CD233	AD	600448	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
Table 4 Diseases of Immune Dysregulation	CD234 deficiency	CD234	AD	600449	P=not attempted	Immunosuppression, alemtuzumab, thymectomy	- IgG subclass may be considered	PMD 20202000
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