

Supplemental Table S4. Pathogenicity of Mutation Based on Clinvar or CADD Score Specific to the N1b PTMCs

Case ID	Hugo symbol	Chromosome	Start position	End position	cDNA change	Protein change	Variant classification	Variants allele frequency	Allelic depth		Clinvar	CADD score
									Reference	Variant		
Case 15	GBA	1	155208310	155208310	c.586A>C	p.K196Q	Missense	0.22	21	6	Pathogenic	24
	PABPC1	8	101724606	101724606	c.956C>T	p.T319I	Missense	0.13	26	4		31
	HECTD4	12	112720990	112720990	c.1020T>G	p.Y340*	Nonsense	0.1	37	3		35
	PPP4R1	18	9570212	9570212	c.1516G>T	p.E506*	Nonsense	0.15	17	3		37
	PTPRC	1	198687262	198687262	c.1490C>A	p.S497*	Nonsense	0.05	123	6		35
	NAPRT1	8	144657683	144657683	c.1201G>T	p.G401W	Missense	0.09	41	4		31
	RAG2	11	36615250	36615250	c.469G>T	p.G157*	Nonsense	0.05	120	6		36
Case 16	WDR65	1	43665068	43665068	c.1436T>C	p.F479S	Missense	0.11	34	4		32
	RYR2	1	237604778	237604778	c.1165C>A	p.R389S	Missense	0.08	46	4		32
Case 17	JAK1	1	65323419	65323419	c.1378G>T	p.E460*	Nonsense	0.14	24	4		41
	ECE2	3	184007313	184007313	c.1817C>A	p.A606D	Missense	0.19	17	4		34
	COL4A3BP	5	74715213	74715213	c.632G>T	p.R211L	Missense	0.12	30	4		32
	TMEM243	7	86848759	86848759	c.61G>T	p.G21W	Missense	0.09	40	4		34
	SLC4A8	12	51853889	51853889	c.1010C>A	p.T337K	Missense	0.1	38	4		34
	CASC3	17	38324163	38324163	c.1712G>A	p.G571D	Missense	0.11	32	4		32
	LRRC4B	19	51021538	51021538	c.1432G>T	p.G478*	Nonsense	0.1	37	4		35
Case 18	ABCC5	3	183660584	183660584	c.3625G>T	p.V1209F	Missense	0.07	43	3		34
	IGF2BP1	17	47119715	47119715	c.1053T>G	p.Y351*	Nonsense	0.1	37	4		34
	ZNF671	19	58232358	58232358	c.1096G>T	p.E366*	Nonsense	0.07	40	3		37
	LAMC3	9	133901682	133901682	c.384T>G	p.Y128*	Nonsense	0.15	76	13		35
	MAPK1	22	22162093	22162093	c.162G>T	p.K54N	Missense	0.14	25	4		32
Case 19	THPO	3	184090840	184090840	c.523C>T	p.R175W	Missense	0.06	46	3		31
	POLE3	9	116172371	116172371	c.116G>C	p.R39P	Missense	0.3	21	9		33
	TCTN3	10	97444373	97444373	c.978T>G	p.Y326*	Nonsense	0.21	19	5		38
Case 20	ADAMTS9	3	64582510	64582510	c.4175G>T	p.G1392V	Missense	0.07	57	4		34
	BRAF	7	140453136	140453136	c.1799T>A	p.V600E	Missense	0.33	10	5	Pathogenic	32
	TG	8	133923660	133923660	c.4041C>A	p.C1347*	Nonsense	0.08	35	3		34
	DDX6	11	118629588	118629588	c.888T>G	p.Y296*	Nonsense	0.25	15	5		38
	ASIC1	12	50474353	50474353	c.1278T>G	p.Y426*	Nonsense	0.1	37	4		36
	PHIP	6	79735777	79735777	c.705T>G	p.Y235*	Nonsense	0.25	60	20	Pathogenic	37
	TAS2R43	12	11243929	11243929	c.900G>A	p.W300*	Nonsense	0.08	61	5		35
Case 21	GMPR	6	16274682	16274682	c.502C>A	p.L168I	Missense	0.06	77	5		33
	SAMD9	7	92733613	92733613	c.1798G>T	p.E600*	Nonsense	0.12	30	4		35
Case 22	GBA	1	155208310	155208310	c.586A>C	p.K196Q	Missense	0.24	18	6	Pathogenic	24
	ANTXR2	4	80929692	80929692	c.1024C>T	p.P342S	Missense	0.06	83	5		31
	NUP98	11	3697601	3697601	c.5191G>A	p.A1731T	Missense	0.08	48	4		32
	TUBGCP3	13	113209227	113209227	c.814G>T	p.E272*	Nonsense	0.13	27	4		37
Case 23	PTN	7	136936120	136936120	c.308T>C	p.F103S	Missense	0.09	41	4		34
	RASGRP2	11	64506875	64506875	c.770G>T	p.R257L	Missense	0.08	46	4		34
	LLGL1	17	18145897	18145897	c.3071A>G	p.D1024G	Missense	0.05	71	4		32

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Supplemental Table S4. Continued

Case ID	Hugo symbol	Chromosome	Start position	End position	cDNA change	Protein change	Variant classification	Variants allele frequency	Allelic depth		Clinvar	CADD score
									Reference	Variant		
Case 24	ZBTB46	20	62407271	62407271	c.982G>T	p.G328*	Nonsense	0.06	46	3		37
	MAP4K4	2	102503568	102503568	c.2950C>A	p.H984N	Missense	0.07	52	4		33
	RBL1	20	35632115	35632115	c.3026G>T	p.S1009I	Missense	0.1	43	5		34
	SIM2	21	38114128	38114128	c.961C>T	p.R321W	Missense	0.07	57	4		33
Case 25	RIPK4	21	43164135	43164135	c.1102G>T	p.G368W	Missense	0.06	45	3		32
	GPBP1L1	1	46124731	46124731	c.29G>T	p.W10L	Missense	0.08	48	4		34
	PCMT1	6	150114743	150114743	c.530C>A	p.S177*	Nonsense	0.1	38	4		40
	HSPA12B	20	3732290	3732290	c.1538C>A	p.P513Q	Missense	0.07	51	4		33
Case 26	USH2A	1	216052393	216052393	c.8271T>G	p.Y2757*	Nonsense	0.13	90	13	Pathogenic	39
	DNMT3A	2	25462084	25462084	c.1654T>C	p.S552P	Splice site	0.07	51	4		32
	DOCK4	7	111395599	111395599	c.4361G>T	p.R1454L	Missense	0.09	42	4		35
	RRM1	11	4141103	4141103	c.821C>A	p.P274Q	Missense	0.08	46	4		34
	OVOL2	20	18022295	18022295	c.394C>T	p.R132C	Missense	0.09	42	4		34
Case 27	CFTR	7	117230409	117230409	c.1682C>A	p.A561E	Missense	0.12	22	3	Pathogenic	33
	PABPC1	8	101724606	101724606	c.956C>T	p.T319I	Missense	0.17	15	3		31
	SLX4	16	3647878	3647878	c.1286G>A	p.R429Q	Missense	0.16	27	5		33
	FANCD2	3	10091090	10091090	c.1446C>A	p.C482*	Nonsense	1.33	26	4		37
	CEP85L	6	118845024	118845024	c.1089G>T	p.W363C	Missense	0.15	22	4		34
	NAPEPLD	7	102760201	102760201	c.764G>T	p.C255F	Missense	0.08	49	4		32
	NXPE2	11	114569423	114569423	c.789T>A	p.C263*	Nonsense	0.06	65	4		35

CADD, Combined Annotation-Dependent Depletion; PTMC, papillary thyroid microcarcinoma.