Supplementary Table 1. Included *FBN1* variants

Variant code	Exon	Nucleotide Change	Predicted Protein Change	Type of *FBN1* variants	Classification of *FBN1* variants	Number of cases	Effect Group	Class	Reference
v1	2	NM_000138.4:c.111delC	p.(Arg38Glufs*70)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v2	2	NM_000138.4:c.2T>A	p.Met1Lys	PTC	PTV	1	Initiation codon variant	P	rs1057516934
v3	3	NM_000138.4:c.239G>A	p.(Cys80Tyr)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs397515767
v4	4	NM_000138.4:c.280T>C	p.(Cys94Arg)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v5	5	NM_000138.4:c.347-2A>G	Exon 5 deletion	IFES	PTV	2	splice-site ±1-2	P	rs1555405056
v6	5	NM_000138.4:c.364C>T	p.(Arg122Cys)	Missense	non-PTV	3	Cys outside cb-EGF or cb-site	LP	rs137854467
v7	5	NM_000138.4:c.385T>G	p.(Cys129Gly)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs199474693
v8	5	NM_000138.4:c.386G>A	p.(Cys129Tyr)	Missense	non-PTV	3	Cys outside cb-EGF or cb-site	LP	rs1566935517
v9	5	NM_000138.4:c.433T>C	p.(Cys145Arg)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs1555405031
v10	5	NM_000138.4:c.400T>G	p.(Cys134Gly)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Attanasio et al. [2]
v11	6	NM_000138.4:c.493C>T	p.(Arg165*)	PTC	PTV	1	Nonsense	P	rs113905529
v12	6	NM_000138.4:c.502T>C	p.(Cys168Arg)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	Jin et al.[3]
v13	7	NM_000138.4:c.718C>T	p.(Arg240Cys)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs137854480
v14	9	NM_000138.4:c.923_926del TCAG	p.(Val308Alafs*21)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v15	9	NM_000138.4:c.939C>G	p.(Cys313Trp)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555401007
v16	9	NM_000138.4:c.953delG	p.(Gly318Valfs*12)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v17	10	NM_000138.4:c.1129T>G	p.(Cys377Gly)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v18	10	NM_000138.4:c.1090C>T	p.(Arg364*)	PTC	PTV	2	Nonsense	P	rs794728165
v19	11	NM_000138.4:c.1285C>T	p.(Arg429*)	PTC	PTV	2	Nonsense	P	rs112645512
v20	12	NM_000138.4:c.1416C>G	p.(Tyr472*)	PTC	PTV	2	Nonsense	P	Takeda et al. [1]
v21	12	NM_000138.4:c.1468+5G>A	Exon 12 deletion	IFES	PTV	1	splice-site non±1-2	LP	rs397515757
v22	13	NM_000138.4:c.1495T>C	p.(Cys499Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	Schrijver et al. [4]
v23	13	NM_000138.4:c.1585C>T	p.(Arg529*)	PTC	PTV	2	Nonsense	P	rs137854476
v24	13	NM_000138.4:c.1477G>T	p.(Glu493*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v25	14	NM_000138.4:c.1623C>A	p.(Cys541*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v26	14	NM_000138.4:c.1637G>T	p.(Cys546Phe)	Missense	non-PTV	4	Cys in cb-EGF	LP	Takeda et al. [1]
v27	14	NM_000138.4:c.1709G>C	p.(Cys570Ser)	Missense	non-PTV	2	Cys in cb-EGF	LP	Ogawa et al. [5]
v28	14	NM_000138.4:c.1693C>T	p.(Arg565*)	PTC	PTV	1	Nonsense	P	rs113422242
v29	14	NM_000138.4:c.1664G>A	p.(Cys555Tyr)	Missense	non-PTV	2	Cys in cb-EGF	LP	rs794728172
v30	15	NM_000138.4:c.1786T>G	p.(Cys596Gly)	Missense	non-PTV	2	Cys in cb-EGF	LP	rs1057520131
v31	16	NM_000138.4:c.1879C>T	p.(Arg627Cys)	Missense	non-PTV	1	Cys in cb-EGF (creating Cys)	LP	rs727503057
v32	16	NM_000138.4:c.1904A>G	p.(Tyr635Cys)	Missense	non-PTV	5	Cys in cb-EGF (creating Cys)	LP	rs1555399816
v33	16	NM_000138.4:c.1910G>A	p.(Cys637Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	Zhao et al. [6]
v34	16	NM_000138.4:c.1949G>A	p.(Arg650His)	Missense	non-PTV	1	Other missense	LP	Takeda et al. [1]
v35	16	NM_000138.4:c.1955G>A	p.(Cys652Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	Comeglio et al. [7]
v36	16	NM_000138.4:c.1911T>G	p.(Cys637Trp)	Missense	non-PTV	1	Cys in cb-EGF	LP	Ogawa et al. [5]
v37	17	NM_000138.4:c.1980delA	p.(Cys661Alafs*57)	PTC	PTV	2	Frameshift	P	Takeda et al. [1]
v38	17	NM_000138.4:c.2025_2026d elTTG	p.(Phe675Valfs*42)	PTC	PTV	1	Frameshift	P	na
v39	17	NM_000138.4:c.2111C>G	p.(Ser704*)	PTC	PTV	1	Nonsense	P	na
v40	17	NM_000138.4:c.2113+3A>C	Exon 17 deletion	IFES	PTV	1	splice-site non±1-2	LP	Ogawa et al. [5]
v41	18	NM_000138.4:c.2153dupC	p.(Ser719Valfs*5)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v42	18	NM_000138.4:c.2121T>G	p.(Tyr707*)	PTC	PTV	1	Nonsense	P	na
v43	19	NM_000138.4:c.2201G>T	p.(Cys734Phe)	Missense	non-PTV	2	Cys in cb-EGF	LP	rs794728187
v44	19	NM_000138.4:c.2237A>G	p.(Tyr746Cys)	Missense	non-PTV	2	Cys in cb-EGF (creating Cys)	LP	rs1555399372
v45	19	NM_000138.4:c.2293+1G>C	Exon 19 deletion	IFES	PTV	1	splice-site ±1-2	P	Ogawa et al. [5]
v46	20	NM_000138.4:c.2413T>G	p.(Cys805Gly)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v47	20	NM_000138.4:c.2413T>C	p.(Cys805Arg)	Missense	non-PTV	2	Cys in cb-EGF	LP	na
v48	20	NM_000138.4:c.2306G>A	p.(Cys769Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs794728190
v49	20	NM_000138.4:c.2302G>T	p.(Glu768*)	PTC	PTV	1	Nonsense	P	na
v50	21	NM_000138.4:c.2447G>T	p.(Cys816Phe)	Missense	non-PTV	2	Cys in cb-EGF	LP	rs397515770
v51	21	NM_000138.4:c.2538_2539+ 5delAGGTATT	Exon 21 deletion	IFES	PTV	1	splice-site ±1-2	P	Takeda et al. [1]
v52	22	NM_000138.4:c.2586T>G	p.(Cys862Trp)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v53	22	NM_000138.4:c.2638G>A	p.(Gly880Ser)	Missense	non-PTV	1	Other missense	LP	rs794728194
v54	22	NM_000138.4:c.2561G>A	p.(Trp854*)	PTC	PTV	1	Nonsense	P	rs1597568968
v55	22	NM_000138.4:c.2651delG	p.(Gly884Glufs*28)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v56	22	NM_000138.4:c.2569_2570d elGT	p.(Val857Hisfs*2)	PTC	PTV	1	Frameshift	P	na
v57	22	NM_000138.4:c.2645C>T	p.(Ala882Val)	Missense	non-PTV	1	Other missense	LP	rs794728195
v58	22	NM_000138.4:c.2581C>T	p.(Arg861*)	PTC	PTV	1	Nonsense	P	rs140583
v59	23	NM_000138.4:c.2722T>C	p.(Cys908Arg)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs1060501021
v60	23	NM_000138.4:c.2702C>G	p.(Ser901*)	PTC	PTV	1	Nonsense	P	na
v61	24	NM_000138.4:c.2729-1G>C	Exon 24 deletion	IFES	PTV	1	splice-site ±1-2	P	Takeda et al. [1]
v62	24	NM_000138.4:c.2753delC	p.(Pro918Glnfs*24)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]

v63	24	NM_000138.4:c.2854G>C	p.(Asp952His)	Missense	non-PTV	1	Other missense	LP	na
v64	25	NM_000138.4:c.2886C>A	p.(Tyr962*)	PTC	PTV	1	Nonsense	P	rs772108557
v65	25	NM_000138.4:c.2953G>A	p.(Gly985Arg)	Missense	non-PTV	2	Other missense	LP	rs794728199
v66	25	NM_000138.4:c.2965_2966delGG	p.(Gly989Tyrfs*2)	PTC	PTV	1	Frameshift	P	na
v67	25	NM_000138.4:c.2942G>A	p.(Cys981Tyr)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	rs727505110
v68	26	NM_000138.4:c.3093G>C	p.(Glu1031Asp)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v69	26	NM_000138.4:c.3095G>A	p.(Cys1032Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs137854481
v70	27	NM_000138.4:c.3209-1G>A	Exon 27 deletion	IFES	PTV	1	splice-site ±1-2	P	Takeda et al. [1]
v71	27	NM_000138.4:c.3302A>G	p.(Tyr1101Cys)	Missense	non-PTV	1	Cys in cb-EGF (creating Cys)	LP	rs1555398625
v72	27	NM_000138.4:c.3337+1G>A	Exon 27 deletion	IFES	PTV	1	splice-site ±1-2	P	rs397515789
v73	28	NM_000138.4:c.3463G>C	p.(Asp1155His)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Stheneur et al. [9]
v74	29	NM_000138.4:c.3559delC	p.(His1187Ilefs*17)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v75	29	NM_000138.4:c.3584G>A	p.(Cys1195Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs886038802
v76	29	NM_000138.4:c.3544T>G	p.(Cys1182Gly)	Missense	non-PTV	1	Cys in cb-EGF	LP	na
v77	29	NM_000138.4:c.3524_3525delTA	p.(Ile1175Argfs*17)	PTC	PTV	1	Frameshift	P	na
v78	30	NM_000138.4:c.3650G>A	p.(Gly1217Asp)	Missense	non-PTV	1	Other missense	LP	rs1555398397
v79	30	NM_000138.4:c.3603C>A	p.(Cys1201*)	PTC	PTV	1	Nonsense	P	Ogawa et al. [5]
v80	30	NM_000138.4:c.3622delT	p.(Cys1208Valfs*22)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v81	30	NM_000138.4:c.3670C>T	p.(Gln1224*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v82	30	NM_000138.4:c.3712G>A	p.(Asp1238Asn)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs794728208
v83	30	NM_000138.4:c.3712+1G>A	Exon 30 deletion	IFES	PTV	1	splice-site ±1-2	P	rs794728209
v84	31	NM_000138.4:c.3713-1G>A	Exon 31 deletion	IFES	PTV	3	splice-site ±1-2	P	Takeda et al.[1]
v85	31	NM_000138.4:c.3746G>C	p.(Cys1249Ser)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs137854458
v86	31	NM_000138.4:c.3781T>G	p.(Tyr1261Asp)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Arbustini et al. [10]
v87	31	NM_000138.4:c.3725G>A	p.(Cys1242Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs137854471
v88	32	NM_000138.4:c.3919T>C	p.(Cys1307Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v89	33	NM_000138.4:c.4021A>C	p.(Asn1341His)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v90	33	NM_000138.4:c.4027G>T	p.(Ala1343Ser)	Missense	non-PTV	1	Other missense	LP	na
v91	33	NM_000138.4:c.4066G>T	p.(Gly1356*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v92	33	NM_000138.4:c.4017T>G	p.(Cys1339Trp)	Missense	non-PTV	1	Cys in cb-EGF	LP	na
v93	34	NM_000138.4:c.4096G>A	p.(Glu1366Lys)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	rs763449629
v94	35	NM_000138.4:c.4313G>A	p.(Ser1438Asn)	Missense	non-PTV	2	Other missense	LP	rs587782945
v95	35	NM_000138.4:c.4280A>G	p.(Tyr1427Cys)	Missense	non-PTV	1	Cys in cb-EGF (creating Cys)	LP	rs1555397548
v96	35	NM_000138.4:c.4331G>T	p.(Cys1444Phe)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v97	35	NM_000138.4:c.4222T>C	p.(Cys1408Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs397515802
v98	36	NM_000138.4:c.4411_4420dupGAGTGTGAGA	p.(Ile1474Argfs*3)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v99	36	NM_000138.4:c.4408T>C	p.(Cys1470Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	Tjeldhorn, et al. [11]
v100	36	NM_000138.4:c.4382G>C	p.(Cys1461Ser)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1057522902
v101	36	NM_000138.4:c.4459G>A	p.(Asp1487Asn)	Missense	non-PTV	1	Other missense	LP	rs113693945
v102	37	NM_000138.4:c.4567C>T	p.(Arg1523*)	PTC	PTV	1	Nonsense	P	rs397515812
v103	37	NM_000138.4:c.4507_4508delGT	p.(Val1503Glnfs*9)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v104	38	NM_000138.4:c.4621C>T	p.(Arg1541*)	PTC	PTV	4	Nonsense	P	rs794728228
v105	38	NM_000138.4:c.4588C>T	p.(Arg1530Cys)	Missense	non-PTV	3	Cys outside cb-EGF or cb-site	LP	rs111401431
v106	38	NM_000138.4:c.4583-1G>A	Exon 38 deletion	IFES	PTV	1	splice-site ±1-2	P	rs1555397176
v107	38	NM_000138.4:c.4615C>T	p.(Arg1539*)	PTC	PTV	1	Nonsense	P	rs111231312
v108	39	NM_000138.4:c.4750G>T	p.(Glu1584*)	PTC	PTV	1	Nonsense	P	Loeys et al. [12]
v109	39	NM_000138.4:c.4786C>T	p.(Arg1596*)	PTC	PTV	1	Nonsense	P	rs113871094
v110	39	NM_000138.4:c.4777G>T	p.(Glu1593*)	PTC	PTV	1	Nonsense	P	Ogawa et al. [5]
v111	40	NM_000138.4:c.4930C>T	p.(Arg1644*)	PTC	PTV	1	Nonsense	P	rs140630
v112	40	NM_000138.4:c.4834G>T	p.(Glu1612*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v113	41	NM_000138.4:c.4973delG	p.(Cys1658Leufs*24)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v114	41	NM_000138.4:c.5060G>A	p.(Cys1687Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v115	41	NM_000138.4:c.4993A>T	p.(Asn1665Tyr)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v116	41	NM_000138.4:c.4973G>A	p.(Cys1658Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555396859
v117	41	NM_000138.4:c.5065+1G>A	Exon 41 deletion	IFES	PTV	2	splice-site ±1-2	P	Ogawa et al. [5]
v118	42	NM_000138.4:c.5177delG	p.(Gly1726Alafs*167)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v119	42	NM_000138.4:c.5162G>A	p.(Cys1721Tyr)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v120	44	NM_000138.4:c.5371T>C	p.(Cys1791Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555396427
v121	44	NM_000138.4:c.5368C>T	p.(Arg1790*)	PTC	PTV	2	Nonsense	P	rs113249837
v122	44	NM_000138.4:c.5372G>A	p.(Cys1791Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs886038848
v123	47	NM_000138.4:c.5672-2A>G	Exon 47 deletion	IFES	PTV	2	splice-site ±1-2	P	rs1060501053
v124	47	NM_000138.4:c.5725_5740delinsCAGTTGAA	p.(Ile1909Glnfs*16)	PTC	PTV	2	Frameshift	P	Takeda et al. [1]
v125	47	NM_000138.4:c.5743C>A	p.(Arg1915Ser)	Missense	non-PTV	1	Other missense	LP	na
v126	47	NM_000138.4:c.5726T>C	p.(Ile1909Thr)	Missense	non-PTV	1	Other missense	LP	rs794728333
v127	47	NM_000138.4:c.5729G>T	p.(Gly1910Val)	Missense	non-PTV	1	Other missense	LP	na
v128	47	NM_000138.4:c.5740T>C	p.(Cys1914Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	na
v129	47	NM_000138.4:c.5788+1G>A	Exon 47 deletion	IFES	PTV	1	splice-site ±1-2	P	rs1555395819

v130	47	NM_000138.4:c.5788+2T>G	Exon 47 deletion	IFES	PTV	1	splice-site ±1-2	P	na
v131	47	NM_000138.4:c.5788+5G>A	Exon 47 deletion	IFES	PTV	2	splice-site non±1-2	LP	rs193922219
v132	47	NM_000138.4:c.5788+5G>C	Exon 47 deletion	IFES	PTV	1	splice-site non±1-2	LP	Takeda et al. [1]
v133	48	NM_000138.4:c.5873G>A	p.(Cys1958Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	Ogawa et al. [5]
v134	48	NM_000138.4:c.5886T>G	p.(Tyr1962*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v135	49	NM_000138.4:c.5966G>T	p.(Cys1989Phe)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1597531796
v136	49	NM_000138.4:c.5950T>C	p.(Cys1984Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555395659
v137	50	NM_000138.4:c.6113G>A	p.(Cys2038Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs363804
v138	51	NM_000138.4:c.6296G>T	p.(Cys2099Phe)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs1131691803
v139	51	NM_000138.4:c.6181T>C	p.(Cys2061Arg)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs1555395267
v140	51	NM_000138.4:c.6169C>T	p.(Arg2057*)	PTC	PTV	2	Nonsense	P	rs763091520
v141	51	NM_000138.4:c.6268G>T	p.(Glu2090*)	PTC	PTV	1	Nonsense	P	na
v142	52	NM_000138.4:c.6379+1G>A	Exon 52 deletion	IFES	PTV	2	splice-site ±1-2	P	rs397515833
v143	53	NM_000138.4:c.6388G>T	p.(Glu2130*)	PTC	PTV	1	Nonsense	P	rs794728334
v144	53	NM_000138.4:c.6448delC	p.(Arg2150Alafs*10)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v145	53	NM_000138.4:c.6487G>T	p.(Glu2163*)	PTC	PTV	1	Nonsense	P	rs1555395191
v146	53	NM_000138.4:c.6388G>A	p.(Glu2130Lys)	Missense	non-PTV	3	Cys outside cb-EGF or cb-site	LP	rs794728334
v147	54	NM_000138.4:c.6518G>A	p.(Gly2173Asp)	Missense	non-PTV	1	Other missense	LP	Ogawa et al. [5]
v148	54	NM_000138.4:c.6577G>T	p.(Glu2193*)	PTC	PTV	1	Nonsense	P	Biggin et al.[13]
v149	54	NM_000138.4:c.6542G>A	p.(Cys2181Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	Waldmuller et al.[14]
v150	55	NM_000138.4:c.6703_6704delGG	p.(Gly2235Ilefs*8)	PTC	PTV	1	Frameshift	P	Ogawa et al. [5]
v151	55	NM_000138.4:c.6665delT	p.(Val2222Glyfs*69)	PTC	PTV	3	Frameshift	P	Ogawa et al. [5]
v152	55	NM_000138.4:c.6658C>T	p.(Arg2220*)	PTC	PTV	1	Nonsense	P	rs113001196
v153	56	NM_000138.4:c.6740-2A>G	Exon 56 deletion	IFES	PTV	2	splice-site ±1-2	P	Takeda et al. [1]
v154	56	NM_000138.4:c.6806T>C	p.(Ile2269Thr)	Missense	non-PTV	3	Other missense	LP	rs193922228
v155	56	NM_000138.4:c.6837delG	p.(Tyr2280Ilefs*11)	PTC	PTV	1	Frameshift	P	Ogawa et al. [5]
v156	56	NM_000138.4:c.6752G>C	p.(Cys2251Ser)	Missense	non-PTV	1	Cys in cb-EGF	LP	na
v157	56	NM_000138.4:c.6748G>A	p.(Glu2250Lys)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs1597520789
v158	57	NM_000138.4:c.6874G>T	p.(Glu2292*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]
v159	57	NM_000138.4:c.6947G>A	p.(Cys2316Tyr)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555394629
v160	57	NM_000138.4:c.6982C>T	p.(Gln2328*)	PTC	PTV	1	Nonsense	P	rs371097218
v161	57	NM_000138.4:c.6997+5G>A	Exon 57 deletion	IFES	PTV	1	splice-site non±1-2	LP	Ogawa et al. [5]
v162	57	NM_000138.4:c.6915delG	p.(Arg 2306 Alafs*92)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v163	58	NM_000138.4:c.7039_7040delAT	p.(Met2347Valfs*19)	PTC	PTV	2	Frameshift	P	rs794728319
v164	58	NM_000138.4:c.7090T>C	p.(Cys2364Arg)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v165	58	NM_000138.4:c.7184G>T	p.(Gly2395Val)	Missense	non-PTV	1	Other missense	LP	rs397515849
v166	58	NM_000138.4:c.7141C>T	p.(Gln2381*)	PTC	PTV	2	Nonsense	P	rs869025414
v167	58	NM_000138.4:c.7015T>G	p.(Cys2339Gly)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Ogawa et al. [5]
v168	58	NM_000138.4:c.7180C>T	p.(Arg2394*)	PTC	PTV	1	Nonsense	P	rs397515848
v169	59	NM_000138.4:c.7266_7267delAG	p.(Gly2423Ilefs*7)	PTC	PTV	1	Frameshift	P	Stheneur et al. [9]
v170	59	NM_000138.4:c.7240C>T	p.(Arg2414*)	PTC	PTV	2	Nonsense	P	rs112550005
v171	59	NM_000138.4:c.7330+3_6delAAGT	Exon 59 deletion	IFES	PTV	1	splice-site non±1-2	LP	Takeda et al. [1]
v172	59	NM_000138.4:c.7327_7330+2delGTAGGT	Exon 59 deletion	IFES	PTV	1	splice-site ±1-2	P	na
v173	60	NM_000138.4:c.7406_7407insTGTT	p.(Cys2470Valfs*19)	PTC	PTV	2	Frameshift	P	Takeda et al. [1]
v174	60	NM_000138.4:c.7339G>A	p.(Glu2447Lys)	Missense	non-PTV	2	Cys outside cb-EGF or cb-site	LP	rs137854464
v175	60	NM_000138.4:c.7408T>C	p.(Cys2470Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555394399
v176	60	NM_000138.4:c.7339G>T	p.(Glu2447*)	PTC	PTV	1	Nonsense	P	Arbustini et al. [10]
v177	61	NM_000138.4:c.7466G>A	p.(Cys2489Tyr)	Missense	non-PTV	3	Cys in cb-EGF	LP	rs1060501077
v178	61	NM_000138.4:c.7565delG	p.(Cys2522Serfs*160)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v179	61	NM_000138.4:c.7545delT	p.(Phe2515Leufs*167)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v180	62	NM_000138.4:c.7582T>C	p.(Cys2528Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1566891701
v181	62	NM_000138.4:c.7664G>T	p.(Gly2555Val)	Missense	non-PTV	1	Other missense	LP	rs1566891654
v182	62	NM_000138.4:c.7636G>A	p.(Gly2546Arg)	Missense	non-PTV	1	Other missense	LP	Bustamante-Aragones et al. [15]
v183	62	NM_000138.4:c.7664delG	p.(Gly2555Aspfs*127)	PTC	PTV	1	Frameshift	P	na
v184	62	NM_000138.4:c.7606G>A	p.(Gly2536Arg)	Missense	non-PTV	1	Other missense	LP	rs397515854
v185	62	NM_000138.4:c.7669+1G>A	Exon 62 deletion	IFES	PTV	1	splice-site ±1-2	P	Baetens et al. [16]
v186	63	NM_000138.4:c.7754T>C	p.(Ile2585Thr)	Missense	non-PTV	9	Other missense	LP	rs727503054
v187	63	NM_000138.4:c.7792dupC	p.(Gln2598Profs*10)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v188	63	NM_000138.4:c.7784G>T	p.(Gly2595Val)	Missense	non-PTV	1	Other missense	LP	na
v189	64	NM_000138.4:c.7831T>C	p.(Cys2611Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v190	64	NM_000138.4:c.7864T>C	p.(Cys2622Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	Takeda et al. [1]
v191	64	NM_000138.4:c.7936T>C	p.(Cys2646Arg)	Missense	non-PTV	1	Cys in cb-EGF	LP	rs1555393863
v192	64	NM_000138.4:c.7938C>A	p.(Cys2646*)	PTC	PTV	1	Nonsense	P	Takeda et al. [1]

v193	64	NM_000138.4:c.7982A>G	p.(Tyr2661Cys)	Missense	non-PTV	1	Cys in cb-EGF (creating Cys)	LP	rs112196241
v194	64	NM_000138.4:c.7906G>T	p.(Gly2636Cys)	Missense	non-PTV	1	Cys in cb-EGF (creating Cys)	LP	Takeda et al. [1]
v195	64	NM_000138.4:c.7828G>A	p.(Glu2610Lys)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs111984349
v196	65	NM_000138.4:c.8063C>G	p.(Ser2688Cys)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	Takeda et al. [1]
v197	65	NM_000138.4:c.8090delC	p.(Pro2697Glnfs*55)	PTC	PTV	1	Frameshift	P	Takeda et al. [1]
v198	65	NM_000138.4:c.8188C>T	p.(Arg2730Trp)	Missense	non-PTV	1	Other missense	LP	na
v199	65	NM_000138.4:c.8135delC	p.(Pro2712Glnfs*40)	PTC	PTV	2	Frameshift	P	Takeda et al. [1]
v200	66	NM_000138.4:c.8377T>G	p.(Tyr2793Asp)	Missense	non-PTV	1	Other missense	LP	na
v201	66	NM_000138.4:c.8521G>T	p.(Glu2841*)	PTC	PTV	1	Nonsense	LP	rs587782948
v202	66	NM_000138.4:c.8378A>G	p.(Tyr2793Cys)	Missense	non-PTV	1	Cys outside cb-EGF or cb-site	LP	rs397515863
v203	66	NM_000138.4:c.8326C>T	p.(Arg2776*)	PTC	PTV	1	Nonsense	LP	rs137854466
CNVs									
v204	20		Exon 20 deletion	IFES	PTV	1	Copy number variation causing exon(s) skipping	P	Takeda et al. [17]
v205	64-66		Exons 64-66 deletion		PTV	1	Copy number variation causing exon(s) skipping	P	na
v206	23-25		Exons 23-25 deletion	IFES	PTV	1	Copy number variation causing exon(s) skipping	P	Takeda et al. [17]
v207	3		Exon 3 deletion	PTC	PTV	1	Copy number variation causing exon(s) skipping	P	Takeda et al. [17]
v208	39-40		Exons 39-40 deletion	IFES	PTV	1	Copy number variation causing exon(s) skipping	P	Takeda et al. [17]
v209	51-63		Exons 51-63 deletion	IFES	PTV	1	Copy number variation causing exon(s) skipping	P	Takeda et al. [17]
v210	1-66		Exons 1-66 deletion		PTV	1	Copy number variation causing exon(s) skipping	P	na
						Total=	278		

PTC, variant creating a premature termination codon; IFES, in-frame exon-skipping; PTV, protein-truncating variant; P, pathogenic; LP, likely-pathogenic; CNV, copy number variation.

<Supplementary Reference>

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