Supplementary Table 1. List of pathogenic clinical variants associated with Mendelian stroke in Korean

Chromosome	Gene	Position	rsID	Reference	Alternative	Protein consequences	Annotation	Clinical significance	CADD score	Allele frequency (gnomAD)	Allele frequency (Korean)
chr16	*ABCC6*	16163087	rs28939701	G	A	p.R1024W	Nonsynonymous SNV	Pathogenic	24.6	0.00007	0.00006
chr16	*ABCC6*	16163159	rs63749794	G	A	p.R1000C	Nonsynonymous SNV	Pathogenic	32.0	0.00040	0.00006
chr16	*ABCC6*	16178950	rs72653787	C	T	p.G641R	Nonsynonymous SNV	Pathogenic	25.6	0.00005	0.00006
chr16	*ABCC6*	16190273	rs779408186	G	C	p.A395G	Nonsynonymous SNV	Likely pathogenic	29.1	0.00004	0.00006
chr12	*ACVRL1*	51913237	rs863223414	G	A	p.R67Q	Nonsynonymous SNV	Pathogenic/likely pathogenic	20.4	.	0.00006
chr12	*ACVRL1*	51915388	rs1085307412	C	G	p.H312Q	Nonsynonymous SNV	Pathogenic	23.8	.	0.00026
chr22	*CECR1*	17181904	rs376785840	T	C	p.Y453C	Nonsynonymous SNV	Pathogenic	23.7	0.00010	0.00013
chr22	*CECR1*	17209538	rs200930463	C	A	p.G47V	Nonsynonymous SNV	Pathogenic/likely pathogenic	22.5	0.00020	0.00013
chr22	*CECR1*	17189980	rs368615054	G	A	p.R71X	Stopgain	Pathogenic	36.0	0.00002	0.00006
chr22	*CECR1*	17189998	rs774963498	G	A	p.R65X	Stopgain	Likely pathogenic	34.0	0.00007	0.00006
chr21	*CBS;CBSL*	43060451	rs769080151	G	A	p.R274W	Nonsynonymous SNV	Likely pathogenic	23.7	0.00006	0.00006
chr21	*CBS;CBSL*	43060475	rs372010465	C	T	p.V266M	Nonsynonymous SNV	Pathogenic/likely pathogenic	24.4	0.00006	0.00006
chr21	*CBS;CBSL*	43060528	rs121964972	G	A	p.T248M	Nonsynonymous SNV	Pathogenic/likely pathogenic	19.0	0.00040	0.00006
chr21	*CBS;CBSL*	43066353	rs121964964	G	A	p.A9V	Nonsynonymous SNV	Pathogenic/likely pathogenic	24.3	0.00040	0.00026
chr2	*COL3A1*	188988627	rs587779592	G	T	p.G207V	Nonsynonymous SNV	Pathogenic	29.0	.	0.00006
chr2	*COL3A1*	188994297	rs587779692	G	A	p.G420S	Nonsynonymous SNV	Pathogenic/likely pathogenic	31.0	.	0.00006
chr13	*COL4A2*	110493246	rs1271683445	G	A	p.G1200S	Nonsynonymous SNV	Likely pathogenic	29.6	0.00001	0.00006
chr19	*COLGALT1*	17560428	rs1478523191	T	G	p.L151R	Nonsynonymous SNV	Pathogenic	31.0	.	0.00019
chr9	*ENG*	127854348	rs1588604597	C	T	p.R3H	Nonsynonymous SNV	Pathogenic	7.5	.	0.00006
chr15	*FBN1*	48470646	rs794728223	C	T	p.G1483R	Nonsynonymous SNV	Likely pathogenic	26.2	.	0.00013
chr15	*FBN1*	48488448	rs137854472	T	C	p.K1043R	Nonsynonymous SNV	Pathogenic	22.2	0.00010	0.00084
chr15	*FBN1*	48489956	rs755477434	A	G	p.C993R	Nonsynonymous SNV	Likely pathogenic	29.5	.	0.00006
chr15	*FBN1*	48470664	rs869025407	C	A	p.E1477X	Stopgain	Pathogenic	42.0	.	0.00006
chr15	*FBN1*	48488480	rs1597564359	G	T	p.C1032X	Stopgain	Pathogenic	37.0	.	0.00006
chrX	*GLA*	101403941	rs781838005	C	G	p.G80A	Nonsynonymous SNV	Likely pathogenic	24.9	.	0.00013
chr11	*HBB*	5226961	rs35890959	C	T	p.V21M	Nonsynonymous SNV	Pathogenic	17.4	.	0.00006
chr10	*HTRA1*	122506817	rs113993970	C	T	p.R302X	Stopgain	Likely pathogenic	40.0	0.00003	0.00006
chr10	*HTRA1*	122508758	rs113993971	C	T	p.R370X	Stopgain	Likely pathogenic	43.0	0.00007	0.00006

https://doi.org/10.5853/jos.2024.03188

Supplementary Table 1. Continued

Chromosome	Gene	Position	rsID	Reference	Alternative	Protein consequences	Annotation	Clinical significance	CADD score	Allele frequency (gnomAD)	Allele frequency (Korean)
chr7	*KRIT1*	92222966	rs886039402	G	A	p.R375X	Stopgain	Pathogenic	37.0	.	0.00006
chr7	*KRIT1*	92234558	rs764960797	G	A	p.R294X	Stopgain	Pathogenic	38.0	0.00003	0.00006
chr17	*NF1*	31201044	rs137854563	T	C	p.L357P	Nonsynonymous SNV	Pathogenic	27.9	.	0.00006
chr17	*NF1*	31214524	rs137854557	A	G	p.Y489C	Nonsynonymous SNV	Pathogenic	24.1	0.00006	0.00006
chr17	*NF1*	31327839	rs786202112	G	A	p.R1849Q	Nonsynonymous SNV	Pathogenic	34.0	.	0.00019
chr17	*NF1*	31357319	rs1060500333	T	G	p.Y2619X	Stopgain	Pathogenic/likely pathogenic	40.0	.	0.00006
chr19	*NOTCH3*	15180173	rs1438626607	G	A	p.R1076C	Nonsynonymous SNV	Pathogenic/likely pathogenic	26.5	.	0.00006
chr19	*NOTCH3*	15187186	rs754554486	G	A	p.R587C	Nonsynonymous SNV	Likely pathogenic	26.2	0.00020	0.00013
chr19	*NOTCH3*	15187315	rs201118034	G	A	p.R544C	Nonsynonymous SNV	Pathogenic/likely pathogenic	23.1	0.00370	0.00109
chr19	*NOTCH3*	15192493	rs145069047	C	G	p.R75P	Nonsynonymous SNV	Pathogenic	20.9	.	0.00051
chr3	*TREX1*	48467514	rs79318303	CTGCTGGCCCCACTGGGT	-	p.P280_A285del	Nonframeshift deletion	Pathogenic/likely pathogenic	.	0.00030	0.00045
chr17	*RNF213*	80385145	rs112735431	G	A	p.R4810K	Nonsynonymous SNV	Conflicting interpretations of pathogenicity	7.4	0.00390	0.02180

CADD, Combined Annotation Dependent Depletion; gnomAD, Genome Aggregation Database; SNV, single nucleotide variant.