

Supplementary Table 1. Demographic information of moyamoya disease patients and normal controls

Case no.	Sex	Age (yr)	Symptoms	Suzuki grade (rt/lt)	Infarct	RNF213 c.14576G>A variant	Use
Moyamoya disease patients							
MMD1	F	4	Headache, TIA (rt)	2/3	Borderzone infarct	G/A	EA, MA, RT, BSP
MMD2	F	17	TIA (lt)	3/2	None	A/A	EA, MA, RT, BSP
MMD3	M	6	TIA (lt)	3/2	None	G/A	EA, MA, RT, BSP
MMD4	M	1	Seizure, hemiparesis (rt)	2/3	Lobar infarct	G/G	RT, PS1
MMD5	F	10	Headache, involuntary movement (lt)	3/4	Lobar infarct	G/G	RT, PS1
MMD6	M	1	Seizure, hemiparesis (lt)	2/2	Lobar infarct, chronic infarct	G/G	RT, PS1
MMD7	F	8	TIA (rt)	3/2	None	G/A	RT, PS1
MMD8	F	13	Headache	2/2	None	G/G	RT, PS2
MMD9	F	11	Headache, lower extremity TIA (both)	4/4	Borderzone infarct	G/A	RT, PS2
MMD10	F	14	TIA (lt)	4/3	None	G/A	RT, PS1
MMD11	F	10	TIA (rt)	4/4	None	G/A	RT, PS1
MMD12	M	7	Headache, TIA (lt)	3/3	None	G/G	PS1
MMD13	M	6	TIA (rt)	1/3	Hemorrhagic infarct	G/G	PS1
MMD14	M	5	TIA (lt)	3/2	None	G/A	PS2
MMD15	M	9	Hemiparesis (lt)	3/3	Lobar infarct	G/A	PS2
MMD16	F	4	TIA (lt)	3/2	None	G/G	PS2
MMD17	M	8	TIA (rt)	1/3	None	G/G	PS2
MMD18	F	4	Hemiparesis (rt)	2/4	Lobar infarct	G/G	PS2
Normal controls							
N1	F	23				G/G	EA, MA, RT, BSP
N2	M	21				G/G	EA, MA, RT, BSP, PS1
N3	M	28				G/G	RT
N4	F	23				G/G	RT, PS2
N5	M	24				G/G	RT, PS1
N6	M	24				G/G	RT, PS1
N7	F	26				G/G	RT, PS1
N8	M	25				G/G	RT, PS1
N9	M	24				G/G	RT, PS1
N10	M	20				G/A	RT, PS1
N11	M	22				G/G	RT, PS1
N12	F	24				G/G	PS2
N13	F	25				G/G	PS2
N14	M	25				G/G	PS2
N15	M	25				G/G	PS2
N16	F	22				G/G	PS2

rt, right; lt, left; RNF213, ring finger protein 213; MMD, moyamoya disease; TIA, transient ischemic attack; G/A, heterozygote (genotype GA); A/A, homozygote (genotype AA); G/G, wild type (genotype GG); EA, expression array; MA, methylation array; RT, reverse-transcription quantitative polymerase chain reaction; BSP, bisulfite sequencing PCR; PS1, pyrosequencing analysis for validation of methylation status on the five candidate; PS2, pyrosequencing analysis for evaluation diagnostic ability of methylation status on *SORT1* gene as a biomarker for MMD; N, normal control.