Supplementary Table 1. Clinical information of MMD patients and healthy volunteers

	Gender	Age (years)	RNF213 variant	Symptom	Infarct	Suzuki grade, right/left	Used	Reference
MMD1	F	7	GA	Headache	No	3/2	Microarray	[11]
MMD2	F	8	Unidentified	TIA	No	3/2	Microarray	[11]
MMD3	M	8	GA	TIA	No	3/3	Microarray	[11]
MMD4	F	19	GA	Weakness	Yes	2/1	Microarray	[11]
MMD5	F	37	GA	TIA	No	3/2	Microarray	[11]
MMD6	F	8	Unidentified	Headache, seizure, TIA	No	3/3	Microarray	[11]
MMD7	F	36	GA	Headache, TIA	No	3/3	Microarray	[11]
MMD8	F	4	GA	Headache	No	3/3	qRT-PCR	[11]
MMD16	M	5	GA	TIA	No	3/2	HCS, qRT-PCR, siRNA transfection (cell viability assay, qRT-PCR, Cell cycle, β-galactosidase assay, Tube formation), Western blot	
MMD17	M	9	GA	TIA	No	3/3	HCS, β-galactosidase assay, Immunofluorescence, qRT-PCR, siRNA transfection (cell viability assay, qRT-PCR, Immunofluorescence)	
MMD18	F	6	GA	TIA	No	2/3	HCS, cell cycle, siRNA transfection (cell viability assay, qRT-PCR, Cell cycle, Tube formation), Western blot	
MMD19	M	10	GA	Headache	No	3/3	HCS, cell cycle, β-galactosidase assay, Tube formation, siRNA transfection (cell cycle, β-galactosidase assay)	
MMD20	M	13	GA	Involuntary movement	No	3/3	HCS, cell cycle	
Healthy volunteers1	M	20	GA	N/A	N/A	N/A	Microarray	[11]
Healthy volunteers2	F	21	GG	N/A	N/A	N/A	Microarray, qRT-PCR	[11]
Healthy volunteers3	F	24	GG	N/A	N/A	N/A	Microarray	[11]
Healthy volunteers4	M	23	GG	N/A	N/A	N/A	Microarray	[11]
Healthy volunteers9	M	22	GG	N/A	N/A	N/A	HCS, cell cycle	
Healthy volunteers10	M	23	GG	N/A	N/A	N/A	HCS, cell cycle	
Healthy volunteers11	F	21	GG	N/A	N/A	N/A	HCS, cell cycle, β-galactosidase assay, Immunofluorescence, Tube formation	
Healthy volunteers12	F	24	GG	N/A	N/A	N/A	HCS, cell cycle, qRT-PCR	
Healthy volunteers13	F	25	GG	N/A	N/A	N/A	HCS, cell cycle, qRT-PCR	

MMD : Moyamoya disease, RNF213 : Ring finger protein 213, F : female, GA : heterozygotes, TIA : transient ischemic attack, M : male, qRT-PCR : real-time quantitative polymerase chain reaction, HCS : high content screening, N/A : not applicable, GG : wild types