Supplementary data:

Genomewide association study identifies no major founder variant in Caucasian moyamoya disease

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Table 1. Primers used for amplification of candidate genes.

Candidate gene	Exon	Forward primer (5′→3′)	Reverse primer (5′→3′)	Annealing T_m
F11R	Exon 1	TTCTCAGCCCTCTAGCTCCAA	GTGAGAGTGGTGAAAGAGGGA	58
	Exon 2,3	GGATCATGGTGACAAAGTGTG	CAAGAGCAAGGACAGAGTGAA	58
	Exon 4, 5	TTGTGGAGGGATTATCATGC	CAAATTGGCTTCCTGCTGAA	58
	Exon 6, 7, 8	ATAGAACCCCAAAAGGTGGGA	AGCGAGAATGCTACAGAAGCA	58
	Exon 9, 10	TTTGAAGCTGAAGTGGGCCTA	TGGGGCACATAGCTGACATTA	58
USF1	Exon 3, 4, 5	ATGACTGGGTTATTTTCCTGGC	TCTTTCAACTCATCCTCCAAGC	58
	Exon 6, 7	ATCCATGTCTCTGAAGGACTTTG	TGCCTGTCCAGGTCTTAATGAT	58
	Exon 8, 9, 10	TCATTAAGACCTGGACAGGCAGT	GAACCTCAGGGAGAGATAAGACTA	58
	Exon 11, 12	TGAGGTCCAAAGTGTGAACCTA	CAAGGACACACCTTCTGAACTT	58
DHX57	Exon 2	CCCAAGACCAGCTCTTTTTA	CAGAAAAGGGTTACAAAGAGGAAG	58
	Exon 4	CAGCAGGTCTGATATAATGAAGTC	AAAGGCAATCCCATGGATGT	58
	Exon 5	TCAGCAACTGAAGAGTAGTCAGGA	CTGAATGACCCCTTCTGATAAGA	58
	Exon 6	GGGGCTTATTGTTATCAGGGAT	TGGGACTTAATGGCTTGTTCAG	58
	Exon 7	GCGTACAGTTGAAACATTCACTGC	AAGTTTGCTGTGGCCTGTAGAA	58
	Exon 8, 9	ATTGCTTAGAGCTACGCTGTCTGA	CCTTATGTCTCAAGGCAGATGT	58
	Exon 10-1, 10-2	CCACCTCTTGGTAATAAGTCAGGA	CCATTACTAGCTGCATGAAACTG	58
	Exon 11	GTAAGGCTTCACATCCAAGAAC	TCTCCCAGCAAGATATGTTACCTC	58
	Exon 12	CTGCCCTTTCCAATTTCTGTT	TAGATGGGGGTCCCAATATGTT	58
	Exon 13	AAGGATGGCATGTGAATTGG	CCCATTGGCAAATATAGTAACA	58
	Exon 14	CAGAGCAAGACCCTGTCTCTAAA	CAAAGCCTGTATTCTTAACCCCA	58
	Exon 15	AGACCACAGGCGAAATTCTC	CTGATTTCCTGCAGCAAATAGG	58
	Exon 16, 17	ACAGGTTGTGGAGAAGTTGCTAA	GGAATGGTAAAGGCAACACTAAGG	58
	Exon 18	TGGTAGAAGTAGAAAACATGAG	GATGTGCCCTAAAAATTCACAGC	58
	Exon 19	TAAGCCTGGGAGATAAAGGTTG	GGTAAGGCCAAACATATTCCAGA	58
	Exon 21	TGAGGAATGAATCCACTTGAGC	GTGCGAAGCAGAAAATCAAGA	58
	Exon 22	CCAACATGGTGAAACCTTGTCT	ACCCGGCCGAAAATGTATACTA	58
	Exon 23	GCATGTGCTTGTTCTGCTTT	TCAATCTTCCCTCAGTCAGTGGTA	58
	Exon 24	TCAGGAAAGTCACGGGCTATAGAT	GCCACCGTCATTCATTTTT	58
	Exon 25	AGGGTTTTTGCTCCTAATGG	TATATACACTGCCTCAGCTGCCTT	58
TNFSF11	Exon 4	AAGCGGTTTATAAGAGTTGGGG	AGACTCAGTTTCCAAGTGCGG	58
	Exon 5	CCTTGTCCTGTGGATGATAGTCA	GCATTCCCTGTAGCTCTTTTACA	58
	Exon 6, 7	CCAATGTTACTATGGCACCTTTG	GGGCTCTCAAAAATTCAGTGAC	58
	Exon 8	CCCCTTCTAGAAAAGAAATGCC	CCATGCCTCTTAGTAGTCTCACA	58

Table 1 (*contd*)

Candidate gene	Exon	Forward primer (5′→3′)	Reverse primer (5′→3′)	Annealing T_m
YTHDF1	Exon 1, 2	ACTGGGACCTGAGAGCGAAA	AAGCTGGCGGAAGAGAGAAA	58
	Exon 3	ATTGAAAGTTTCCCGTCCC	ACTGATGCCCAAGGTTAACA	58
	Exon 5	ACAGCTTTCTTTCTGGAAGGA	TGCCAATGTCCATGTTATGC	58
		AACGGTGGGACAAATGTGAA	ATGTGCCGGAGCTGGTTATT	58
		TACCTGCTCTTCAGCGTCAAT	CAGTACTCTCCGCAGGGAAA	58
	Exon 6	TGTGGCTTGGGTACCATTCA	GGGATGAGAAATATGAAGTCTCACA	58
BIRC7	Exon 1	AGTTCCTTCCCGTCTTGTTCA	ACCACTGCTCCGTCATCA	58
	Exon 2	TACAAGTGACATCCCCCACCAA	TCACAGGAAAAGTGGGTACCT	58
	Exon 3, 4, 5	AGGTACCCACTTTTCCTGTGA	TCTGCACATCCTGCCCATCT	58
	Exon 6	TCTGGATCCTGTTATGCTGTG	TAAAAACAAGCTGACGCTGC	58
NKAIN4	Exon 1	CTCCAAGCCAAGGAATCTCTG	CGAATCTCAGAATCGGAACTTG	58
	Exon 3	TCGTCAAACAAAGGTGTGGT	TTCTAGATGTCCAGGCAGCAAA	58
	Exon 4	TTTCTCCATGCTGTGCATC	ACAGAGCCATGCACAGACTGA	58
	Exon 5	GTATTCCTTTCCCCACCATG	TGATACCTTAACCTCTCCAGCAAG	58
	Exon 6	TTGAGAGCTGAGGATACCCAC	TCTGCTAATCTCTTCCCTGGTGT	58
	Exon 7	CTCTGCTGTCATGTCAGGGCTT	CTGGCATGAAGCAGATGTTAGA	58
	Exon 8	TTTCTGCCAAAGGCCTGAAG	TGGTGTCCAGTACTTAGAACCCA	58
ARFGAP1	Exon 2	TCCTTCAGTAAGTGGTCCCTG	CACTGATGAGCGCCAGAAAT	58
	Exon 3, 4	CGTCTTCCCATCTGAACATTT	GGTGTGTCAGAACCAGCAGAA	58
	Exon 5	TTGTAAAGTGAGTCCTGGCCT	TGACTTCTGGGAAAAGAGCTG	58
	Exon 6	GGCATTGCTGGGGTTTCA	TGGGACTCACTGCAATCAGA	58
	Exon 7	TGGCTTTGGGGACTTGCT	AAAACGTCGCTGAAGGTAAGG	58
	Exon 8	TTTCTGTTGTGCCCTGGGT	TGTTCCTGCAGAACCTCCAA	58
	Exon 9	CAGCATTTGTGCTAATCCAGT	ACCAGCTACCAGTGAACAGGA	58
	Exon 10-1	TCGCGCAGTTTCTGAAGGT	TGATGCAGATCCTGGATCTTA	58
	Exon 10-2	TTCCTGCAAGAATTGGTGG	AGGCATCATTCCCATACCAGA	58
	Exon 10-3, 10-4	TGTAGAACTTTCTGGGGCTCA	AAGAGCTCCCACCACATGCTT	58
		TTTGCTGCCATCAGTCCCA	CAGCAAAGATGCCAGCCACCT	58
	Exon 12	TCTGAAGCGGGAGAAACAGAT	TCACACTGCAGTTGCTGATTC	58

Table 2. Summary of the demographic and clinical profiles of the MMD cases and the unrelated controls.

Items	Cases	Controls
No. of participants	38	41
Male:female	14:24	8:33
Age at participation (years ± SD[a])	31.37 ± 14.83	27.34 ± 11.55
Family history of MMD	3	–
Range of age at onset	9 months–58 years	–
Mean age at onset (years ± SD)	22.93 ± 16.95	–
Young onset (<15 year)	14	–
Adult onset (≤15 year)	24	–
Clinical symptoms		
Cerebral infarction	18	–
TIA[b]	6	–
Stroke	3	–
Headache	4	–
Hemorrhage	1	–
Epilepsy	2	–
Asymptomatic	1	–
Other[c]	3	–
Clinical status		
Bilateral	35	–
Unilateral	3	–

[a]SD, standard deviation; [b]TIA, transient ischemic attack; [c]other, includes dizziness, haemichorea and motor retardation.

Table 3. Associations of the top significant SNPs in the GWAS.

dbSNP id	Locus	Alleles (risk allele)	Position (build 36.3)*	Function	Gene	DD	Dd	dd	Minor allele frequency	DD	Dd	dd	Minor allele frequency	OR (95% CI)	*P*
Case–control						Cases				Controls					
rs1023115	1q23.3	C/T (C)	159201443	Unknown	–	11	25	2	0.62	3	17	21	0.28	6.63 (2.54–17.29)	2.60E-06
rs11681583	2p22.1	A/G (G)	38983439	Unknown	–	10	23	5	0.57	1	17	23	0.23	6.51 (2.56–16.57)	2.93E-06
rs3742257	13q14.11	C/T (C)	42071198	Intron	*TNFSF11*	2	19	17	0.30	18	18	5	0.66	0.20 (0.09–0.45)	5.73E-06
rs2058364	17p13.3	G/T (T)	5557413	Unknown	–	15	18	5	0.63	1	22	18	0.29	5.57 (2.32–13.39)	5.90E-06
rs720607	20q13.33	C/T (T)	61366354	Unknown	–	3	15	20	0.28	12	26	3	0.61	0.17 (0.07–0.42)	5.59E-06
Case-only						Cases with cerebral infarction				Cases without cerebral infarction					
rs17078141	3p22.1	G/A (A)	40334981	Unknown	–	0	3	15	0.08	3	14	3	0.50	0.04 (0.01–0.23)	6.10E-06
rs2178763	4q22.3	A/G (A)	97819016	Unknown	–	7	10	1	0.67	0	8	12	0.20	21.81 (2.75–172.70)	6.88E-06

*Human reference genome (NCBI build 36.3); d, major allele; D, risk allele or minor allele; OR, odds ratio; CI, confidence interval.

Table 4. Summary of variants detected of the candidate genes by direct sequencing.

Gene description	dbSNP ID	Position (build 36.3)*	Effect	Frequency of alternative allele in Caucasian	Reference sequence	Case 1	Case 2	Case 3	Case 4	Case 5	Control 1
F11R	rs2481085	159257530	5′ UTR	0.306	CC	CC	CT	CC	CT	CT	CC
NM_016946.4	rs28365978	159257502	5′ UTR	0.542	GG	AA	GA	AA	GA	GA	GG
1q21.2-q21.3	rs28365979	159257500	5′ UTR	0.558	GG	TT	GT	TT	GT	GT	GG
	rs78350026	159257499	5′ UTR	0.558	AA	GG	GA	GG	GA	GA	AA
	rs2297219	159237779	IVS1–12	0.292	CC	CC	CG	CC	CG	CG	CC
	rs790055	159237770	IVS1–3	0.832	TT	CC	CC	CC	CC	CC	TT
	rs2297220	159237639	IVS2+60	0.322	CC	CC	CG	CC	CG	CG	CC
	rs2297221	159237592	IVS2+107	0.279	CC	CC	CT	CC	CT	CT	CC
	rs869985	159236554	IVS5+6	0.279	TT	TT	TG	TT	TT	TG	TT
	rs790056	159236209	IVS6-48	0.832	GG	AA	AA	AA	AA	AA	GG
	rs2297222	159235734	IVS8+76	0.352	AA	AA	AG	AA	AG	AG	AA
	rs67708418	159235633^159235634	IVS8+228^229	1.000	–/–	insG/insG	insG/insG	insG/insG	insG/insG	insG/insG	–/–
	rs41269993	159235233	3′ UTR	0.319	CC	CC	CT	CC	CT	CT	CC
USF1	rs2516839	159279745	Exon 3 UTR	0.700	GG	GA	AA	GA	GA	GA	GG
NM_007122.3	rs2073653	159279384	IVS3–88	0.086	AA	AG	AA	AG	AG	AG	AA
1q22-q23	rs2073655	159279214	IVS4+33	0.325	CC	CT	CT	CC	CT	CT	CC
	rs2774276	159278340	IVS6–80	0.783	CC	GG	GG	GG	GG	GG	CC
	rs2073657	159277415	IVS8–69	0.674	GG	GA	AA	GA	GA	GA	GG
	rs2516841	159277398	IVS8–112	0.783	TT	CC	CC	CC	CC	CC	TT
	rs2073658	159277386	IVS8–100	0.283	GG	GA	GA	GG	GA	GA	GG
DHX57	rs3099999	38944200	IVS3–35	1.000	AA	GG	GG	GG	GG	GG	GG
NM_198963.1	rs1131752	38937796	IVS7–6	0.383	TT	TT	TT	TC	CC	TT	TC
2p22.1	rs10219	38937544	Exon 8 UTR	0.358	AA	AA	AA	AG	GG	AA	AG
	rs7598922	38935848	**N587S**	0.560	AA	GG	GG	AA	AA	GG	AG
	rs11124651	38928860	IVS11+55	0.514	TT	GG	GG	TG	TT	GG	TG
	rs7589162	38907049	IVS16+113	0.433	CC	CG	GG	CC	CC	GG	CG
	rs3770681	38903645	S1095S	0.603	TT	TG	GG	TT	TT	GG	TG
	rs34582261	38903568	IVS18+71	0.934	AA	delA/delA	delA/delA	delA/delA	delA/delA	delA/delA	delA/delA
	rs3112138	38896143	IVS21+24	0.983	CC	TT	TT	TT	TT	TT	TT
	rs7419838	38894349	IVS22+118	0.400	CC	CC	CC	CT	TT	CC	CT
	rs13029406	38883578	IVS23–17	0.552	GG	GA	AA	GA	GG	AA	GA
	rs1530852	38879136	IVS24–48	0.568	TT	TC	TT	TC	TT	TT	TC
	rs1530853	38879059	C1349C	0.558	CC	CT	CC	CT	CC	CC	CT
TNFSF11	rs9533157	42046313	Exon 4 UTR	0.983	CC	GG	GG	GG	GG	GG	GG
NM_033012.3	rs2296533	42046565	Exon 4 UTR	0.500	TT	TC	CC	TT	TT	CC	TT
13q14	rs2277438	42053168	IVS4–91	0.847	GG	AA	AA	AA	GA	AA	GA
	rs2277439	42053443	IVS5+14	0.847	GG	AA	AA	AA	GA	AA	GA
	rs3742258	42078576	IVS7–57	0.033	AA	AC	AA	AA	AA	AC	AA
	rs9562415	42079024	F235F	0.033	TT	TC	TT	TT	TT	TC	TT
YTHDF1	–	61317802	5′ near gene	–	CC	CT	CC	CC	CC	CT	CC

Table 4 (*contd*)

Gene description	dbSNP ID	Position (build 36.3)*	Effect	Frequency of alternative allele in Caucasian	Reference sequence	Case 1	Case 2	Case 3	Case 4	Case 5	Control 1
NM_017798.3	rs6090310	61317513	IVS1+150	0.408	GG	CC	CC	CC	CC	CC	CC
20q13.33	rs45518532	61317501	IVS1+162	0.054	GG	GA	GG	GG	GG	GA	GA
	rs6122103	61305140	A199A	0.261	CC	CT	CC	CC	CT	TT	CT
	rs41282994	61304537	D400D	0.092	CC	CT	CC	CC	CC	CT	CT
	rs6122394	61303959	IVS5+122	0.203	CC	CC	CC	CC	CT	CT	CC
BIRC7	rs3810467	61337651	5′ near gene	0.194	GG	GG	GG	GA	GA	GA	GG
NM_139317.1	rs6011691	61337899	G2G	0.100	AA	AG	AA	AA	AA	AG	AG
20q13.33	rs6011692	61338273	IVS1+31	0.069	GG	GA	GG	GG	GG	GA	GA
	rs139816557	61339616	IVS1-84	0.013	AA	AA	AA	AA	AT	AA	AA
	rs6062855	61339668	IVS1-32	0.425	TT	CC	CC	TC	TC	TC	CC
	rs6010878	61340052	IVS2-141	0.558	CC	AA	AA	AA	CA	AA	AA
	rs2273487	61340271	S176S	0.491	CC	TT	TT	CT	CT	CT	TT
	rs744590	61340520	IVS4+109	0.403	CC	TT	TT	CT	CT	CT	TT
	rs744591	61340612	Exon 5 UTR	0.389	CC	AA	AA	CA	CA	CA	AA
	rs744592	61340665	IVS5+26	0.403	TT	CC	CC	TC	TC	TC	CC
	rs1075557	61340910	IVS5-49	0.467	AA	GG	GG	AG	AG	AG	GG
	rs3215649	61341053^61341054	Exon 6 UTR	1.000	–/–	insG/insG	insG/insG	insG/insG	–/insG	insG/insG	insG/insG
	rs1075556	61341079	Exon 6 UTR	0.361	AA	GG	GG	AG	AG	GG	GG
	rs1077019	61341172	**E223Q**	0.133	GG	GG	GG	GC	GG	GG	GG
NKAIN4	rs6090334	61356478	5′ near gene	0.858	CC	GG	CC	GG	GG	GG	CC
NM_152864.3	rs35796278	61356332	5′ UTR	–	CC	GG	GG	GG	GG	GG	GG
20q13.33	rs149519714	61356025	IVS1+169	0.042	CC	CC	CC	CG	CC	CC	CC
	rs34203226	61355897	IVS1+297	0.483	CC	CA	CC	CA	CA	AA	CA
	rs927081	61351741	Y61Y	0.514	TT	TC	CC	CC	TC	TT	TC
	rs2273497	61350719	IVS3-27	0.852	TT	CC	CC	CC	TC	CC	CC
	rs1129659	61350614	**K91Q**	0.425	AA	AC	CC	AC	AC	AA	AC
	rs78017705	61350518	IVS4+94	0.067	CC	CT	CC	CC	CC	TT	CT
	rs2236194	61349454	**A131D**	0.150	CC	CC	CC	CA	CC	CC	CC
	rs872808	61349395	**C151G**	0.408	TT	TG	GG	TG	TG	TT	TG
	rs41282998	61345958	IVS5-77	0.097	TT	TC	TT	TT	TT	CC	TC
	rs2236186	61345942	IVS5-61	0.567	TT	GG	GG	GG	TG	GG	GG
	rs183907681	61345701	IVS6+120	0.003	AA	AA	AC	AA	AA	AA	AA
	–	61345645^61345644	IVS6+176^177	–	–/–	insA/insA	–/–	–/–	–/–	–/–	–/–
	rs1047826	61343350	IVS7-47	0.583	TT	CC	CC	CC	TC	CC	CC
ARFGAP1	rs6011712	61377520	IVS2+93	0.431	TT	TT	TT	TC	TC	CC	TC
NM_175609.1	rs2236195	61383160	IVS8+16	0.542	TT	TT	TT	TC	TC	CC	TC
20q13.33	rs11086161	61385820	Exon 10 UTR	0.375	GG	TT	TT	GT	GT	GG	GT
	rs13042357	61386824	Exon 10 UTR	0.442	CC	TT	TT	CT	CC	CT	CT
	rs6090342	61387686	Exon 10 UTR	1.000	AA	GG	GG	GG	GG	GG	GG

*Human reference genome (NCBI build 36.3). UTR, untranslated region; IVS, intervening sequence; del, deletion; ins, insertion. Allele frequencies were retrieved from HapMap Project or 1000 Genomes Project. Missense variants are indicated in bold.

Table 5. Association of SNPs in *RNF213* and *ACTA2* with MMD.

dbSNP ID	Locus	Alleles (risk allele)	Position (build 36.3)*	Function	Gene	Cases			Minor allele frequency	Controls			Minor allele frequency	OR (95% CI)	*P*
						DD	Dd	dd		DD	Dd	dd			
rs9915508	17q25.3	G/T (G)	75845351	unknown	*RNF213*	10	18	10	0.50	10	20	11	0.49	1.05 (0.57–1.93)	0.880
rs7219131	17q25.3	C/T (C)	75867365	intron	*RNF213*	5	22	11	0.42	9	22	10	0.49	0.73 (0.37–1.44)	0.368
rs7220465	17q25.3	A/G (A)	75874606	intron	*RNF213*	2	10	26	0.18	6	20	15	0.39	0.36 (0.17–0.77)	0.005
rs11869363	17q25.3	A/G (G)	75881354	intron	*RNF213*	1	14	23	0.21	7	21	13	0.43	0.33 (0.15–0.72)	0.003
rs12937242	17q25.3	A/G (A)	75882889	intron	*RNF213*	1	7	30	0.12	3	16	22	0.27	0.38 (0.16–0.90)	0.019
rs9905727	17q25.3	A/G (G)	75887409	intron	*RNF213*	1	9	28	0.14	2	18	21	0.27	0.44 (0.19–1.03)	0.049
rs8066993	17q25.3	C/T (T)	75894625	intron	*RNF213*	1	12	25	0.18	3	21	17	0.33	0.42 (0.19–0.93)	0.027
rs8081176	17q25.3	C/T (C)	75898582	intron	*RNF213*	3	17	18	0.30	9	20	12	0.46	0.50 (0.25–0.98)	0.037
rs9674807	17q25.3	C/T (T)	75900772	intron	*RNF213*	2	16	20	0.26	9	17	15	0.43	0.50 (0.25–0.97)	0.036
rs7222014	17q25.3	A/G (A)	75908064	3′ UTR	*RNF213*	5	24	9	0.45	9	16	16	0.41	1.15 (0.6–2.18)	0.673
rs8082521	17q25.3	A/C (C)	75916752	unknown	*RNF213*	2	24	12	0.37	7	18	16	0.39	0.90 (0.45–1.79)	0.762
rs4890008	17q25.3	G/T (T)	75920214	unknown	*RNF213*	0	5	33	0.07	0	1	40	0.01	6.06 (0.67–54.48)	0.063
rs10782008	17q25.3	A/G (G)	75920466	unknown	*RNF213*	4	25	9	0.43	7	19	15	0.40	1.17 (0.59–2.31)	0.662
rs8074015	17q25.3	A/G (A)	75920875	unknown	*RNF213*	2	24	12	0.37	7	18	16	0.39	0.90 (0.45–1.79)	0.762
rs7211876	17q25.3	C/T (T)	75925955	unknown	*RNF213*	1	15	22	0.22	2	13	26	0.21	1.11 (0.51–2.41)	0.798
rs4890010	17q25.3	A/G (G)	75930774	intron	*RNF213*	3	26	9	0.42	7	19	15	0.40	1.10 (0.55–2.21)	0.794
rs11150856	17q25.3	C/T (C)	75937477	intron	*RNF213*	0	9	29	0.12	2	8	31	0.15	0.80 (0.32–1.96)	0.616
rs8067292	17q25.3	C/T (T)	75948435	intron	*RNF213*	10	21	7	0.54	9	14	18	0.39	1.74 (0.94–3.23)	0.073
rs4078429	17q25.3	A/G (G)	75949967	intron	*RNF213*	0	2	36	0.03	0	4	37	0.05	0.51 (0.09–2.98)	0.447
rs8070106	17q25.3	A/G (G)	75959041	intron	*RNF213*	10	21	7	0.54	9	15	17	0.40	1.68 (0.90–3.12)	0.096
rs6565681	17q25.3	A/G (A)	75963089	intron	*RNF213*	1	20	17	0.29	1	12	28	0.17	2.27 (0.97–5.29)	0.052
rs7224239	17q25.3	A/G (G)	75969771	intron	*RNF213*	4	19	15	0.36	11	22	8	0.54	0.44 (0.22–0.89)	0.017
rs8072774	17q25.3	A/G (A)	75977649	missense	*RNF213*	0	11	27	0.14	0	4	37	0.05	3.77 (1.08–13.12)	0.028
rs3185057	17q25.3	A/G (A)	75978442	synonymous	*RNF213*	0	4	34	0.05	1	7	33	0.11	0.47 (0.14–1.57)	0.200
rs8359	17q25.3	C/T (T)	75982646	3′ UTR	*RNF213*	0	11	27	0.14	0	4	37	0.05	3.77 (1.08–13.12)	0.028

Table 5 (*contd*)

dbSNP id	Locus	Alleles (risk allele)	Position (build 36.3)*	Function	Gene	Cases			Minor allele frequency	Controls			Minor allele frequency	OR (95% CI)	*P*
						DD	Dd	dd		DD	Dd	dd			
rs1372325	10q23.31	A/G (A)	90683549	missense	*ACTA2*	0	8	30	0.11	2	10	29	0.17	0.58 (0.23–1.47)	0.242
rs17446483	10q23.31	C/T (T)	90683685	intron	*ACTA2*	1	9	28	0.14	0	7	34	0.09	1.83 (0.66–5.05)	0.238
rs12571023	10q23.31	G/T (T)	90685964	intron	*ACTA2*	0	4	34	0.05	0	6	35	0.07	0.69 (0.18–2.65)	0.582
rs2119685	10q23.31	A/G (G)	90687675	intron	*ACTA2*	3	8	27	0.18	2	12	27	0.20	0.94 (0.45–1.96)	0.872
rs17114283	10q23.31	A/G (G)	90698439	intron	*ACTA2*	0	1	37	0.01	0	0	41	0.00	–	0.224
rs12241898	10q23.31	A/G (A)	90700955	intron	*ACTA2*	0	0	38	0.00	0	0	41	0.00	–	–
rs1441734	10q23.31	C/T (T)	90706817	unknown	*ACTA2*	8	19	11	0.46	13	15	13	0.50	0.87 (0.48–1.56)	0.642
rs7907084	10q23.31	C/T (T)	90709217	unknown	*ACTA2*	5	21	12	0.41	10	16	15	0.44	0.88 (0.48–1.64)	0.698
rs1926203	10q23.31	G/T (G)	90717314	unknown	*ACTA2*	4	14	20	0.29	3	18	20	0.29	0.98 (0.50–1.95)	0.965
rs7088326	10q23.31	A/G (A)	90718761	unknown	*ACTA2*	7	19	12	0.43	11	15	15	0.45	0.94 (0.52–1.70)	0.840
rs11202914	10q23.31	C/T (T)	90720191	unknown	*ACTA2*	4	21	13	0.38	7	16	18	0.37	1.07 (0.56–2.04)	0.838
rs12765241	10q23.31	A/G (A)	90730481	unknown	*ACTA2*	1	10	27	0.16	0	7	34	0.09	2.05 (0.74–5.65)	0.155
rs4934433	10q23.31	A/C (C)	90730933	unknown	*ACTA2*	4	16	18	0.32	6	13	22	0.30	1.05 (0.56–1.97)	0.890
rs10887876	10q23.31	C/T (T)	90735110	unknown	*ACTA2*	0	9	29	0.12	1	9	31	0.13	0.86 (0.33–2.25)	0.763
rs983751	10q23.31	G/T (T)	90736673	unknown	*ACTA2*	0	12	26	0.16	1	9	31	0.13	1.23 (0.49–3.08)	0.661
rs4934434	10q23.31	A/C (A)	90737149	unknown	*ACTA2*	5	21	12	0.41	6	16	19	0.34	1.33 (0.69–2.55)	0.388
rs3758483	10q23.31	A/G (G)	90738716	5′ near gene	*ACTA2*	0	12	26	0.16	1	9	31	0.13	1.23 (0.49–3.08)	0.661
rs3740286	10q23.31	C/T (C)	90741320	intron	*ACTA2*	5	20	13	0.39	5	16	20	0.32	1.41 (0.73–2.72)	0.308
rs7097467	10q23.31	C/T (C)	90743224	intron	*ACTA2*	0	9	29	0.12	1	9	31	0.13	0.86 (0.33–2.25)	0.763
rs1926196	10q23.31	A/G (A)	90743728	intron	*ACTA2*	10	22	6	0.55	10	17	14	0.45	1.50 (0.80–2.83)	0.204

*Human reference genome (NCBI build 36.3); d, major allele; D, risk allele or minor allele; OR, odds ratio; CI, confidence interval. UTR, untranslated region. A nominal association ($P < 0.002$ for *RNF213* and $P < 0.0025$ for *ACTA2*) was considered to be the threshold for significance.

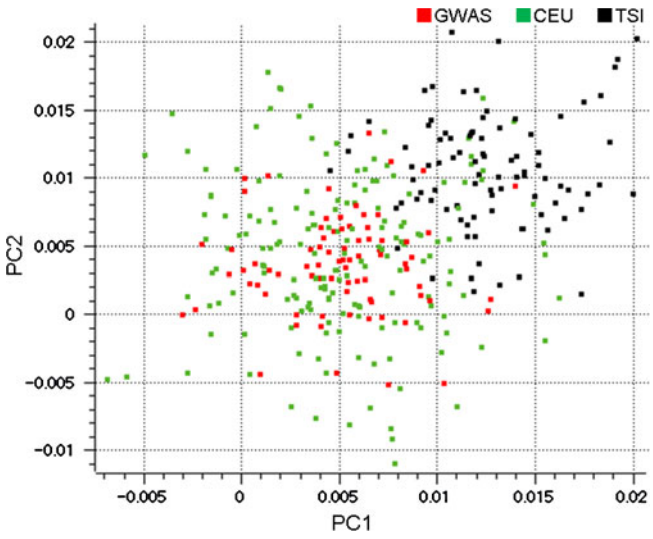

Figure 1. Principal component analysis for genetic matching of the 79 individuals in the present study and unrelated individuals from the HapMap Project. Plot of the first two principal components (PC1 and PC2) from the Caucasian GWAS samples and the HapMap three samples. Population descriptors: GWAS, Caucasian participants in this study; CEU, residents of UT, USA with northern and western European ancestry from the CEPH collection; TSI, Tuscan in Italy.

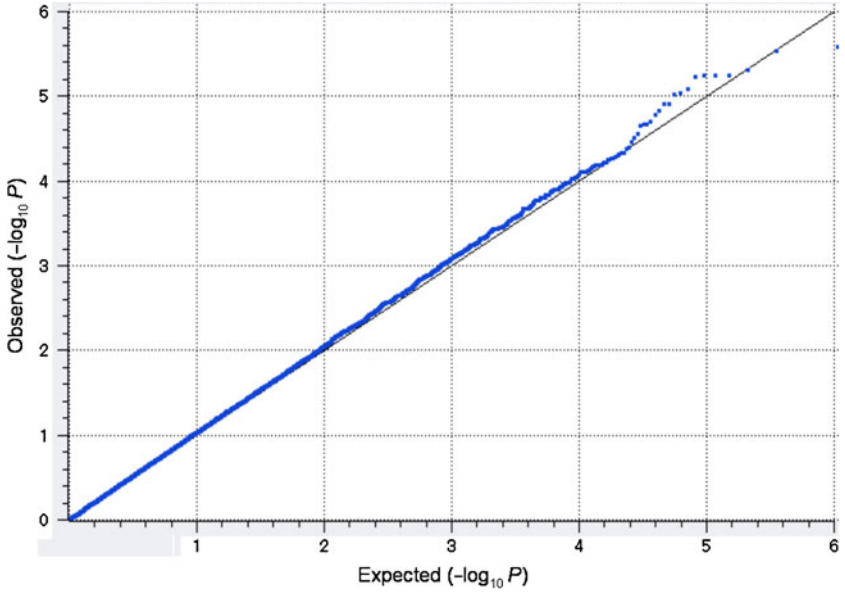

Figure 2. Genomewide quantile–quantile (Q–Q) plot. The x-axis represents the expected $-\log10\,P$ and the y-axis represents the observed $-\log10\,P$. Blue indicates the P values for the SNPs that passed the quality control criteria. The black diagonal line represents $y = x$.