

Supplementary information for

Quadruplex-Single Nucleotide Polymorphisms (Quad-SNP) influence gene expression difference among individuals

Aradhita Baral et al. 2011

List of SNPs as present in Figure 1A

CEU	CHB	JPT	YRI
rs3758076	rs9258795	rs854773	rs2256820
rs356968	rs3752129	rs709117	rs8051991
rs2256820	rs7034648	rs3784400	rs1673396
rs9970563	rs3784400	rs3755799	rs3743742
rs477935	rs3755799	rs12753665	rs11659635
rs1673396	rs2294213	rs1122634	rs10413334
rs11659635	rs11570094	rs12529490	rs8055206
rs8051991	rs1022479	rs3752129	rs2285659
rs2725391	rs7272495	rs7272495	rs9970563
rs9319617	rs7099117	rs2002259	rs3751068
rs3751068	rs12753665	rs2294213	rs3759541
rs3743742	rs2002259	rs9258795	rs1022479
rs2285659	rs1871043	rs12531377	rs4821599
rs3744028	rs9450638	rs9450638	rs4140871

rs1573736	rs3862629	rs3862629	rs9450638
rs3784400	rs12672074	rs1871043	rs9258795
rs9450638	rs2303115	rs4140871	rs3752129
rs9258795	rs1122634	rs4360516	rs3827249
rs4821599	rs4140871	rs2256820	rs9968886
rs4140871	rs12529490	rs3751068	rs2303115
rs12753665	rs4821599	rs356968	rs3784400
rs11570094	rs356968	rs477935	rs12672074
rs1122634	rs2285659	rs1673396	rs7272495
rs2303115	rs477935	rs1573736	rs7957096
rs1022479	rs3758076	rs11659635	rs3758076
rs7272495	rs12440651	rs3758076	rs2269432
rs1871043	rs9348724	rs9319617	
rs2002259	rs1673396	rs9970563	
rs3752129	rs7579147	rs7579147	
rs2294213	rs459091	rs9348724	
rs3862629	rs3743742	rs2285659	
rs459091	rs11659635	rs8051991	
rs12672074	rs9970563	rs3744028	
	rs4360516	rs4821599	
	rs9319617	rs2303115	
	rs1633	rs12672074	
	rs3744028	rs1022479	
	rs1573736	rs459091	
	rs8051991	rs3743742	
	rs3751068		
	rs2256820		

Supplementary Table1

Details of 54 Quad-SNP significantly correlated(Stranger et al.2007) with gene expression

Distance_from_TSS	Gene	SNP	Population	P value	Position of SNP
24	PLCB2	rs3784400	CEU-CHB-JPT-YRI	1.0E-12	Upstream of TSS
91	FLJ10640	rs3743742	CEU-CHB-JPT-YRI	5.0E-09	Upstream of TSS
94	PLTP	rs2294213	JPT	1.2E-06	Downstream of TSS
94	PLTP	rs2294213	CEU-CHB-JPT	1.8E-11	Downstream of TSS
94	PLTP	rs2294213	CHB-JPT	6.8E-09	Downstream of TSS
94	PLTP	rs2294213	CHB-JPT	9.6E-11	Downstream of TSS
212	LOC221711	rs9348724	JPT	2.8E-07	Downstream of TSS
212	LOC221711	rs9348724	CHB-JPT	7.1E-08	Downstream of TSS
231	ZNF175	rs3752129	CEU-CHB-JPT-YRI	2.5E-09	Downstream of TSS
514	CP110	rs2285659	CEU-CHB-JPT-YRI	5.9E-09	Downstream of TSS
556	LOC114984	rs8055206	YRI	6.7E-07	Upstream of TSS
2096	Hs.485895	rs9450638	CEU	2.3E-09	Upstream of TSS
2096	Hs.485895	rs9450638	CHB	2.3E-09	Upstream of TSS
2096	Hs.485895	rs9450638	JPT	1.1E-10	Upstream of TSS
2096	Hs.485895	rs9450638	CEU-CHB-JPT-YRI	7.8E-17	Upstream of TSS
2096	Hs.485895	rs9450638	CEU-CHB-JPT	7.7E-26	Upstream of TSS
2096	Hs.485895	rs9450638	CHB-JPT	2.0E-17	Upstream of TSS
3190	FLJ21347	rs1122634	JPT	8.3E-08	Upstream of TSS
3190	FLJ21347	rs1122634	CEU	4.2E-10	Upstream of TSS
3190	FLJ21347	rs1122634	CHB	1.4E-14	Upstream of TSS
3190	FLJ21347	rs1122634	CEU-CHB-JPT	7.6E-19	Upstream of TSS

3190	FLJ21347	rs1122634	CHB-JPT	2.7E-20	Upstream of TSS
3190	FLJ21347	rs1122634	YRI	3.6E-09	Upstream of TSS
5993	ZFP100	rs10413334	YRI	1.7E-14	Downstream of TSS
6224	STXBP2	rs2303115	CEU-CHB-JPT-YRI	3.4E-08	Downstream of TSS
7435	hmm32897*	rs12531377	CHB	4.6E-09	Upstream of TSS
7435	hmm32897*	rs12531377	JPT	1.7E-08	Upstream of TSS
8787	IVD*	rs2289332	CHB	4.6E-07	Upstream of TSS
9835	Hs.43687	rs7034648	CHB	2.8E-07	Downstream of TSS
10495	C1QTNF6	rs4140871	CEU-CHB-JPT-YRI	5.7E-11	Upstream of TSS
10495	C1QTNF6	rs4140871	CEU-CHB-JPT	1.1E-08	Upstream of TSS
11386	Hs.473973	rs3827249	YRI	1.7E-08	Upstream of TSS
12182	MRPL28	rs1573736	JPT	2.5E-06	Upstream of TSS
12182	MRPL28	rs1573736	CEU-CHB-JPT	3.6E-06	Upstream of TSS
12182	MRPL28	rs1573736	CHB-JPT	1.8E-07	Upstream of TSS
13312	ARHGEF19	rs4360516	CHB-JPT	8.3E-09	Upstream of TSS
13733	LOC114984	rs8051991	CEU-CHB-JPT-YRI	2.0E-09	Downstream of TSS
14016	TRIM47	rs3744028	CEU-CHB-JPT	3.3E-07	Upstream of TSS
14250	C11orf17	rs3751068	YRI	2.7E-09	Downstream of TSS
14250	C11orf17	rs3751068	YRI	6.6E-08	Downstream of TSS
14250	C11orf17	rs3751068	CEU-CHB-JPT-YRI	7.4E-08	Downstream of TSS
14250	C11orf17	rs3751068	CEU-CHB-JPT-YRI	1.3E-07	Downstream of TSS
14547	Hs.6637	rs11570094	CEU	5.5E-07	Downstream of TSS
14547	Hs.6637	rs11570094	CHB	4.4E-08	Downstream of TSS
16233	C1QTNF6	rs4821599	CEU-CHB-JPT-YRI	1.1E-10	Upstream of TSS
16233	C1QTNF6	rs4821599	CEU-CHB-JPT	5.5E-08	Upstream of TSS
16261	TCL6	rs3759541	YRI	1.8E-15	Downstream of TSS
18986	TCL1B	rs3759541	YRI	5.5E-13	Downstream of TSS

20665	LOC284184	rs9319617	CEU	9.6E-15	Upstream of TSS
20665	LOC284184	rs9319617	CEU-CHB-JPT	1.3E-22	Upstream of TSS
20665	LOC284184	rs9319617	CHB-JPT	4.5E-09	Upstream of TSS
20665	LOC284184	rs9319617	CEU	9.6E-15	Upstream of TSS
20681	Hs.519979	rs356968	YRI	8.8E-16	Upstream of TSS
20681	LOC284184	rs2725391	CEU	2.0E-16	Downstream of TSS
22387	C6orf64	rs9968886	YRI	2.4E-08	Downstream of TSS
26069	KIAA1271	rs7272495	CEU-CHB-JPT-YRI	5.1E-18	Downstream of TSS
26069	KIAA1271	rs7272495	CEU-CHB-JPT	3.4E-12	Downstream of TSS
26069	KIAA1271	rs7272495	CHB-JPT	4.1E-07	Downstream of TSS
31502	hmm28069	rs7579147	CHB-JPT	7.1E-07	Upstream of TSS
33965	PHC2	rs9970563	CEU-CHB-JPT-YRI	5.1E-12	Downstream of TSS
33965	PHC2	rs9970563	CEU-CHB-JPT	3.8E-10	Downstream of TSS
38632	MRPL21	rs477935	JPT	4.0E-13	Upstream of TSS
38632	MRPL21	rs477935	CEU-CHB-JPT	3.8E-09	Upstream of TSS
38632	MRPL21	rs477935	CHB-JPT	3.1E-13	Upstream of TSS
39391	LOC389362	rs1633	CHB	2.3E-08	Upstream of TSS
39391	LOC389362	rs1633	JPT	9.3E-08	Upstream of TSS
47209	C10orf64	rs7099117	CHB-JPT	4.1E-09	Downstream of TSS
48036	ZNRD1	rs11754620	JPT	7.4E-09	Upstream of TSS
55477	VRK3	rs10413334	YRI	2.2E-06	Downstream of TSS
64717	LOC347981	rs356968	CHB-JPT	3.3E-12	Downstream of TSS
64717	LOC347981	rs356968	JPT	1.0E-11	Downstream of TSS
64717	LOC347981	rs356968	CEU-CHB-JPT	2.8E-09	Downstream of TSS
67263	SIAT4C	rs3862629	CEU-CHB-JPT	4.4E-09	Downstream of TSS
73430	LOC197322	rs2002259	CEU-CHB-JPT	2.8E-14	Downstream of TSS
76773	RABGEF1	rs12531377	JPT	1.8E-09	Downstream of TSS

78314	LOC347981	rs9258795	JPT	4.7E-10	Upstream of TSS
78314	LOC347981	rs9258795	CEU-CHB-JPT-YRI	5.0E-11	Upstream of TSS
78314	LOC347981	rs9258795	CEU-CHB-JPT	1.2E-08	Upstream of TSS
79999	ARL6IP4	rs7957096	YRI	4.9E-07	Downstream of TSS
81210	Hs.211939	rs4846233	CHB	9.5E-07	Downstream of TSS
88929	ZNRD1	rs2256820	JPT	1.9E-07	Upstream of TSS
88929	ZNRD1	rs2256820	CEU-CHB-JPT-YRI	7.3E-10	Upstream of TSS
88929	ZNRD1	rs2256820	CHB-JPT	1.3E-07	Upstream of TSS
104427	C1orf16	rs12753665	CEU	7.4E-09	Downstream of TSS
104427	C1orf16	rs12753665	CEU-CHB-JPT	3.2E-07	Downstream of TSS
104916	PKHD1L1	rs1673396	CEU	3.7E-09	Downstream of TSS
104916	PKHD1L1	rs1673396	CHB	2.7E-11	Downstream of TSS
104916	PKHD1L1	rs1673396	JPT	1.0E-08	Downstream of TSS
104916	PKHD1L1	rs1673396	YRI	3.4E-07	Downstream of TSS
104916	PKHD1L1	rs1673396	CEU-CHB-JPT-YRI	5.7E-28	Downstream of TSS
104916	PKHD1L1	rs1673396	CEU-CHB-JPT	5.6E-26	Downstream of TSS
104916	PKHD1L1	rs1673396	CHB-JPT	6.8E-19	Downstream of TSS
116532	FLJ20635	rs1022479	JPT	3.0E-08	Downstream of TSS
116532	FLJ20635	rs1022479	CEU-CHB-JPT-YRI	9.4E-14	Downstream of TSS
116532	FLJ20635	rs1022479	CEU-CHB-JPT	3.5E-17	Downstream of TSS
116532	FLJ20635	rs1022479	CHB-JPT	7.2E-12	Downstream of TSS
120642	FCGBP	rs459091	CEU-CHB-JPT	1.3E-08	Upstream of TSS
120642	FCGBP	rs459091	CHB-JPT	5.7E-09	Upstream of TSS
130039	PIGN	rs11659635	JPT	1.7E-07	Downstream of TSS
130039	PIGN	rs11659635	CEU-CHB-JPT-YRI	1.3E-10	Downstream of TSS
130039	PIGN	rs11659635	CEU-CHB-JPT	9.8E-09	Downstream of TSS
178647	RPS6KB2	rs1871043	CEU-CHB-JPT	1.2E-07	Downstream of TSS

211988	SHMT1	rs854773	JPT	8.4E-07	Downstream of TSS
213067	CCNDBP1	rs12440651	CHB	2.9E-06	Upstream of TSS
217820	CA13	rs3758076	CEU-CHB-JPT-YRI	1.1E-06	Downstream of TSS
240123	FLJ12442	rs3755799	CHB-JPT	4.2E-07	Upstream of TSS
267493	LOC402476	rs12672074	YRI	4.5E-08	Downstream of TSS
267493	LOC402476	rs12672074	CEU-CHB-JPT-YRI	3.3E-18	Downstream of TSS
267493	LOC402476	rs12672074	CEU-CHB-JPT	4.9E-12	Downstream of TSS
267493	LOC402476	rs12672074	CHB-JPT	5.5E-10	Downstream of TSS
272678	HEMK	rs2269432	YRI	1.3E-07	Upstream of TSS
327324	SUPT3H	rs12529490	CHB-JPT	2.2E-09	Downstream of TSS
327324	SUPT3H	rs12529490	JPT	4.3E-10	Downstream of TSS

* gene nearest to the Quad-SNP

Supplementary Table 2

Details of 5 Quad- SNPs for which CD and melting experiments are given in text (Figure 1B). SNPs are shown in red font.

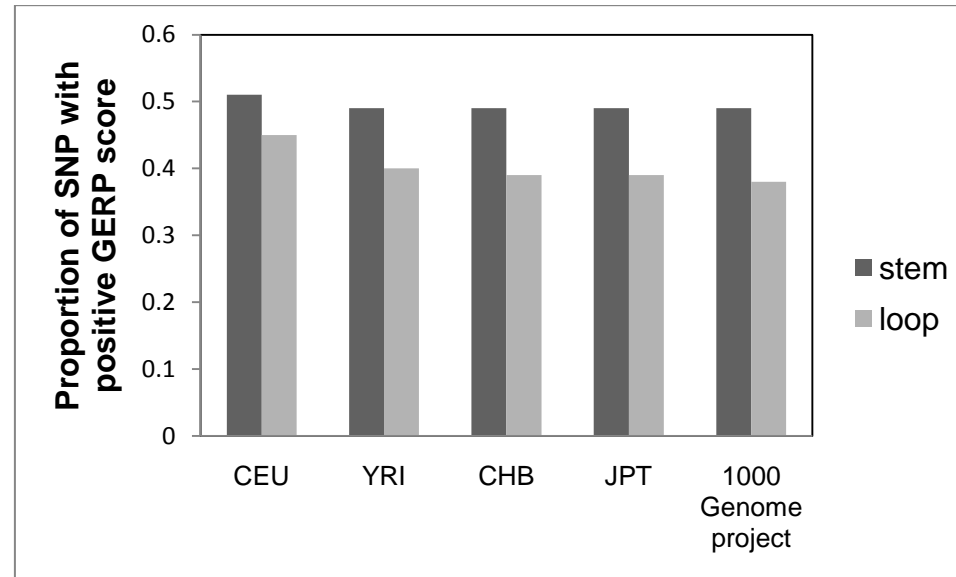
SNP id	Gene	G4 sequence with 1st allelic form	G4 sequence with 2nd allelic form
rs37521129	ZNF175	GGGGGTTTCTGGGGTGGGTGATGGGTG	AGGGGTTTCTGGGGTGGGTGATGGGTG
rs9450638	Hs485895	GGGGCTGAAGGGCTAAGGGAAAAGGGGA	GGGGCTGAAGGGCTAAGGGAAAAGGGGA
rs11570094	Hs.6637 - S	GGGGTGGAGGAGGGAAAGGGCCTTTTGGG	GGGGTGGAGGAGGGAAAGGGCCTTTTGGG
rs3744028	TRIM47	GGGCAGGGATGTGGGGAGGCAGGG	GGGTCAGGGATGTGGGGAGGCAGGG
rs477935	MRLP21	GGGGTGCTGGGCAGGGCAGTGGG	GGGGTGCCGGGCAGGGCAGTGGG

Supplementary Table 3

Derived allele frequency (DAF) of Quad-SNP was binned into 10 categories. Fraction of Quad-SNPs present in each category of DAF across the four HapMap populations is shown. For each bin the fraction has been calculated with respect to the number of Quad-SNP present in respective regions.

	Fraction of SNP											
	CEU			YRI			CHB			JPT		
DAF	STEM	LOOP	QUAD-SNP	STEM	LOOP	QUAD-SNP	STEM	LOOP	QUAD-SNP	STEM	LOOP	QUAD-SNP
0-0.1	0.48	0.38	0.436	0.48	0.39	0.441	0.44	0.37	0.408	0.46	0.40	0.431
0.1-0.2	0.12	0.08	0.103	0.10	0.12	0.108	0.13	0.12	0.124	0.10	0.08	0.093
0.2-0.3	0.08	0.05	0.069	0.07	0.07	0.072	0.05	0.07	0.060	0.06	0.07	0.068
0.3-0.4	0.07	0.06	0.069	0.07	0.11	0.086	0.07	0.06	0.064	0.05	0.09	0.068
0.4-0.5	0.04	0.11	0.069	0.08	0.09	0.082	0.04	0.06	0.050	0.08	0.08	0.082
0.5-0.6	0.07	0.05	0.058	0.08	0.08	0.079	0.06	0.10	0.074	0.03	0.05	0.036
0.6-0.7	0.05	0.10	0.072	0.04	0.03	0.036	0.05	0.06	0.053	0.04	0.06	0.050
0.7-0.8	0.02	0.05	0.031	0.02	0.04	0.029	0.03	0.06	0.046	0.03	0.03	0.028
0.8-0.9	0.03	0.05	0.038	0.02	0.05	0.032	0.01	0.04	0.025	0.04	0.07	0.053
0.9-1	0.04	0.08	0.055	0.05	0.02	0.036	0.13	0.06	0.096	0.11	0.07	0.093

Supplementary Figure 1



Higher proportion of stem Quad-SNP harbor positive RS score by GERP analysis as compared to loop SNPs (two tailed t-test, $P = 0.001$).