

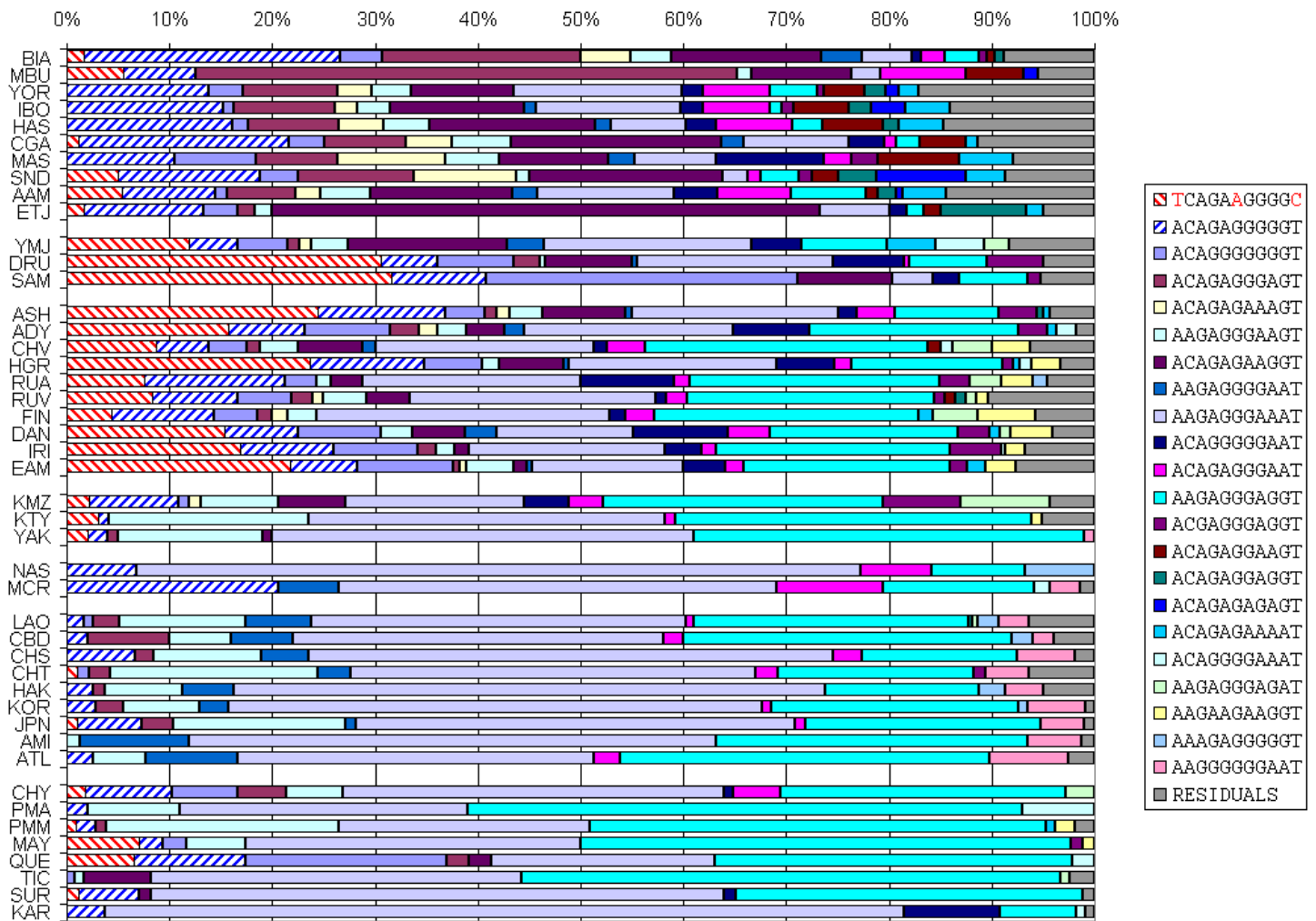
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**Supplemental Data**

**The Distribution and Most Recent Common Ancestor  
of the 17q21 Inversion in Humans**

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**Figure S1. Eight H1 Polymorphic SNPs and Three Inversion-Marking SNPs**



This figure shows the data from figure 4 divided into populations instead of regional averages. Like in figure 4 the haplotypes presented here are composed of eight H1 polymorphic SNPs that have a difference in frequency between Africa and Southwest Asia (rs242924, rs242940, rs171440, 173365, rs171443, rs4616318, rs962885, and rs242933), as well as three inversion marker SNPs (rs11079718, rs1396862, and rs1078830; the H2 alleles are red in the legend). The red striped bar represents the inversion. The inversion is most similar to the blue striped haplotype (differing at only the three inversion marker SNPs). The blue striped haplotype is more common in Africa compared to Southwest Asia. In Africa the blue-striped haplotype ranges in frequencies from 7% to 25%, whereas in Southwest Asia the frequencies range 4.7% to 9.2%.

Table S1. SNPs Typed

		UCSC	Computed	Number			H2			Used in	Typed	Populations
SNP		Absolute	Distance	of			Allele	Used in	Used in	MCRA	in	other than
Number	dbSNP #	Position(bp) <sup>a</sup>	in bp to	Populations	Assay	SNP	Rare <sup>c</sup>	Figure 4	Figure 5	Haplotypes	NHP	46 Std <sup>d</sup>
			next SNP	Typed	Type	Lineage <sup>b</sup>						
1	rs1724436	41,013,675	4,633	63 pops	Illumina	H1					*	1,2,3,4,5
2	rs1724427	41,018,308	13,506	63 pops	Illumina	H2		*			*	1,2,3,4,5
3	rs16970596	41,031,814	38,642	63 pops	Illumina	H1	*				*	1,2,3,4,5
4	rs241039	41,070,456	11,011	62 pops	Taqman	inv marker				*	*	2,3,4,5
5	rs434428	41,081,467	2,692	49 pops	Taqman	inv marker				*	*	6
6	rs417968	41,084,159	3,497	63 pops	Illumina	H1						1,2,3,4,5
7	rs1635302	41,087,656	3,605	63 pops	Illumina	H1					*	1,2,3,4,5
8	rs241027	41,091,261	8,866	46 pops	Taqman	inv marker				*	*	
9	rs1635298	41,100,127	5,828	63 pops	Illumina	H1					*	1,2,3,4,5
10	rs1630095	41,105,955	1,741	63 pops	Illumina	H1					*	1,2,3,4,5
11	rs1635291	41,107,696	3,958	63 pops	Illumina	H1					*	1,2,3,4,5
12	rs1724400	41,111,654	5,985	63 pops	Illumina	H1					*	1,2,3,4,5
13	rs2049515	41,117,639	3,601	49 pops	Taqman	inv marker				*		6
14	rs16941049	41,121,240	2,316	63 pops	Illumina	H1					*	1,2,3,4,5
15	rs7215239	41,123,556	5,351	63 pops	Illumina	H1					*	1,2,3,4,5
16	rs10491144	41,128,907	4,189	49 pops	Taqman	inv marker				*	*	6
17	rs1724422	41,133,096	3,792	63 pops	Illumina	H1					*	1,2,3,4,5
18	rs968028	41,136,888	642	46 pops	Taqman	H1					*	
19	rs1724425	41,137,530	1,593	63 pops	Illumina	H1					*	1,2,3,4,5
20	rs1526123	41,139,123	888	63 pops	Illumina	H1					*	1,2,3,4,5
21	rs9912362	41,140,011	2,008	63 pops	Illumina	H1						1,2,3,4,5
22	rs8073838	41,142,019	2,560	63 pops	Illumina	H1					*	1,2,3,4,5
23	rs1880748	41,144,579	7,745	63 pops	Illumina	H1					*	1,2,3,4,5
24	rs1880750	41,152,324	6,430	46 pops	Taqman	H1	*				*	
25	rs10514879	41,158,754	3,954	63 pops	Illumina	inv marker					*	1,2,3,4,5
26	rs2902662	41,162,708	1,002	46 pops	Taqman	inv marker				*	*	
27	rs1880753	41,167,031	1,637	63 pops	Illumina	H1					*	1,2,3,4,5
28	rs12938476	41,168,668	13,769	63 pops	Illumina	H1					*	1,2,3,4,5
29	rs1880756	41,182,437	13,286	63 pops	Illumina	H1					*	1,2,3,4,5
30	rs11079718	41,195,723	6,771	46 pops	Taqman	inv marker			*		*	
31	rs6503449	41,202,494	8,428	63 pops	Illumina	H1					*	1,2,3,4,5
32	rs4074461	41,210,922	72	63 pops	Illumina	H1					*	1,2,3,4,5
33	rs4074462	41,210,994	4,174	63 pops	Illumina	H1	*				*	1,2,3,4,5
34	rs12942300	41,215,168	10,745	46 pops	Taqman	H1					*	
35	rs7209436	41,225,913	6,878	46 pops	Taqman	H1					*	
36	rs4792887	41,232,791	3,027	63 pops	Illumina	H1					*	1,2,3,4,5
37	rs110402	41,235,818	5,329	46 pops	Taqman	botH		*			*	
38	rs242924	41,241,147	7,233	63 pops	Illumina	H1			*		*	1,2,3,4,5
39	rs242940	41,248,380	887	63 pops	Illumina	H1			*		*	1,2,3,4,5

		UCSC	Computed Distance	Number of			H2			Used in	Typed	Populations
SNP		Absolute	in bp to	Populations	Assay	SNP	Allele	Used in	Used in	MCRA	in	other than
Number	dbSNP #	Position(bp) <sup>a</sup>	next SNP	Typed	Type	Lineage <sup>b</sup>	Rare <sup>c</sup>	Figure 4	Figure 5	Haplotypes	NHP	46 Std <sup>d</sup>
40	rs171440	41,249,267	4,882	63 pops	Illumina	H1			*		*	1,2,3,4,5
41	rs242937	41,254,149	2,706	63 pops	Illumina	botH		*			*	1,2,3,4,5
42	rs173365	41,256,855	1,923	46 pops	Tagman	H1			*		*	
43	rs1396862	41,258,778	10,547	46 pops	Tagman	inv marker			*		*	
44	rs242948	41,269,325	10,353	63 pops	Illumina	botH		*			*	1,2,3,4,5
45	rs171443	41,279,678	4,912	46 pops	Tagman	H1			*		*	
46	rs4616318	41,284,590	6,830	46 pops	Tagman	H1			*		*	
47	rs962885	41,291,420	2,839	63 pops	Illumina	H1			*		*	1,2,3,4,5
48	rs242933	41,294,259	7,642	63 pops	Illumina	H1			*		*	1,2,3,4,5
49	rs1078830	41,301,901	1,700	46 pops	Tagman	inv marker		*	*		*	
50	rs916793	41,310,477	9,619	55 pops	Tagman	inv marker		*			*	1,2,3,4,
51	rs4792891	41,329,294	6,474	63 pops	Illumina	H1					*	1,2,3,4,5
52	rs930119	41,335,768	5,191	63 pops	Illumina	H1					*	1,2,3,4,5
53	rs2280004	41,340,959	8,245	63 pops	Illumina	H1					*	1,2,3,4,5
54	rs3785880	41,349,204	3,185	63 pops	Illumina	H1					*	1,2,3,4,5
55	rs767057	41,354,650	6,102	46 pops	Tagman	H1					*	
56	rs16940742	41,360,752	14,821	63 pops	Illumina	H1					*	1,2,3,4,5
57	rs242557	41,375,573	7,026	46 pops	Tagman	H1					*	
58	rs242562	41,382,599	4,652	46 pops	Tagman	H1					*	
59	rs878917	41,387,251	8,276	63 pops	Illumina	H1					*	1,2,3,4,5
60	rs17650901	41,395,527	9,638	49 pops	Tagman	inv marker		*			*	6
61	E_TAUBSAHI_10	41,405,165	1,499	46 pops	Tagman	H1	*				*	
62	rs9896485	41,406,664	1,018	63 pops	Illumina	H1					*	1,2,3,4,5
63	rs1800547	41,407,682	78	50 pops	Tagman	H1	*				*	2
64	rs17651213	41,407,760	1,524	46 pops	Tagman	inv marker					*	
65	rs2435214	41,424,176	5,549	42 pops	Tagman	H1					*	
66	rs1052553	41,429,725	85	46 pops	Tagman	inv marker					*	
67	rs1991556	41,439,239	4,098	46 pops	Tagman	H1					*	
68	C_2560324_10	41,443,337	4,226	46 pops	Tagman	H1					*	
69	E_TAUIVS11_10	41,447,563	7,079	46 pops	Tagman	inv marker					*	
70	rs916896	41,454,642	2,766	63 pops	Illumina	H1					*	1,2,3,4,5
71	rs9468	41,457,408	3,834	63 pops	Tagman	inv marker					*	1,2,3,4,5
72	rs7521	41,461,242	4,448	46 pops	Tagman	H1					*	
73	rs1078997	41,465,690	7,000	63 pops	Illumina	H1					*	1,2,3,4,5
74	C_2560314_10	41,472,690	3,146	46 pops	Tagman	H1					*	
75	rs10514899	41,475,836	8,141	63 pops	Illumina	H1					*	1,2,3,4,5
76	rs12150447	41,483,977	3,164	59 pops	Tagman	inv marker					*	1,3,4,5
77	rs4792827	41,487,141	4,725	63 pops	Illumina	H1					*	1,2,3,4,5
78	rs933317	41,491,866	5,301	46 pops	Tagman	H1					*	
79	rs2838	41,497,167	2,869	63 pops	Tagman	inv marker					*	1,2,3,4,5

SNP		UCSC	Computed Distance	Number of	Assay	SNP	H2	Used in	Used in	Used in	Typed in	Populations other than
Number	dbSNP #	Absolute Position(bp) <sup>a</sup>	in bp to next SNP	Populations Typed	Type	Lineage <sup>b</sup>	Allele Rare <sup>c</sup>	Figure 4	Figure 5	MCRA Haplotypes	NHP	46 Std <sup>d</sup>
80	rs2301732	41,500,036	11,514	63 pops	Illumina	H1					*	1,2,3,4,5
81	rs2066899	41,511,550	33,300	46 pops	Taqman	H1					*	
82	rs7225002	41,544,850	7,082	63 pops	Illumina	H1					*	1,2,3,4,5
83	rs1468241	41,551,932	10,511	46 pops	Taqman	inv marker					*	
84	rs4792831	41,562,443	1,646	63 pops	Illumina	H1					*	1,2,3,4,5
85	rs6503457	41,564,089	12,142	63 pops	Illumina	H1					*	1,2,3,4,5
86	rs1528075	41,576,231	7,169	46 pops	Taqman	inv marker					*	
87	rs2696578	41,583,400	9,102	63 pops	Illumina	H1					*	1,2,3,4,5
88	rs1528072	41,592,502	11,774	58 pops	Taqman	inv marker					*	2,5
89	rs2240758	41,604,276	11,191	63 pops	Illumina	H1					*	1,2,3,4,5
90	rs2240756	41,615,467	404,011	63 pops	Illumina	H1					*	1,2,3,4,5
	total coverage bp/snp	601,792 6686.577778										

<sup>a</sup> NCBI Build 36.1

<sup>b</sup> Whether the SNP is polymorphic on H1 and/or H2 or is an inversion marker

<sup>c</sup> The allele that is found on the H2 lineage is rare on the H1 lineage

<sup>d</sup> 1=ZAR 2=SOM, NMK, HAZ, MOH 3=GRK, TOS 4=SRD, RMJ 5=LIS, KWT, THT, KCH, PNG, SMN, MLY, GHB 6= CAT, SPB, PLA

**Table S2. Non-Human Primate Alleles of Inversion Marking SNPs**

	NHP Consensus	Gorilla gorilla	Hylobates	Pan paniscus	Pan troglodytes	Pongo pygmaeus
H1 alleles	13	11	10	11	11	8
H2 alleles	7	6	3	5	6	9
unknown	1	4	8	5	4	4

**Table S3. Non-Human Primate Alleles of SNPs Polymorphic only on H1**

	NHP Consensus	Gorilla gorilla	Hylobates	Pan paniscus	Pan troglodytes	Pongo pygmaeus
H2 Allele	55	54	39	56	57	38
non-H2 Allele	4	5	7	4	4	6
unknown	3	3	16	2	1	18

**Table S4. STRPS Primers**

STRP	forward primer	reverse primer
MAPT07	TCAGATTCGGTTTCGGGGGTCGTTATCG	GCTGGAGGGTCTTCACCACCAGATTCGC
MAPT08	CAATGAGCCAAGATCACACCTACTG	GGGTCCTATCATTCTCTCTGCTATCA
MAPT09	TTCTTCTTGCCAATAGCAAGGATAC	CAGTATATACCCAGAAAAGTGCAATGTG
MAPT14	CTCACAAATGTTCTCTGTCTCTTCTGTC	GATGCATCTAATTTAAGGCATCTTATGA

**Table S5. Relationships between HGDP H2 Individuals**

Population	Ind. 1	Ind. 1 Genotype	Ind. 2	Ind. 2 Genotype	Relationship <sup>a</sup>
Mandenka	HGDP00914	Heterozygous	HGDP00919	Heterozygous	HS or AV
Palestinian	HGDP00681	H1 Homozygous	HGDP00684	Heterozygous	HS or AV
Palestinian	HGDP00682	H1 Homozygous	HGDP00743	Heterozygous	HS or AV
Palestinian	HGDP00723	H2 Homozygous	HGDP00743	Heterozygous	HS or AV
Bedouin	HGDP00616	Heterozygous	HGDP00633	Heterozygous	PO
Bedouin	HGDP00617	H2 Homozygous	HGDP00635	H2 Homozygous	HS or AV
Orcadian	HGDP00794	Heterozygous	HGDP00801	Heterozygous	PO
Balochi	HGDP00082	Heterozygous	HGDP00084	H2 Homozygous	FS
Kalash	HGDP00321	Heterozygous	HGDP00326	H1 Homozygous	HS, AV, CO, or GG

<sup>a</sup>HS- Half Sibling, AV- Avuncular, PO- Parent-Offspring, FS- Full Sibling, CO- Cousin, GG- Grandparent-Grandchild