

Supplementary Table 1: Genotyping data for 3 SNP and 3 microsatellite markers at the *JPH3* locus in HDL2 positive individuals and families

Individual	Origin	<i>JPH3</i> expansion	Marker											
			D16S3074		D16S3048		(SNP1) rs2544638		(SNP2) rs3849254		(SNP3) rs918368		D16S413	
			Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2
HC49	SA Black	+	16	23	25	24	C	C	T	C	C	T	14	25
HC51	SA Black	+	12	23	25	22	C	C	T	C	C	C	20	24
HC71	SA Black	+	12	12	25	24	C	C	T	C	C	C	20	23
HC77	SA Black	+	12	16	25	24	C	C	T	T	C	T	20	26
HC86	SA Black	+	12	16	25	20	C	C	T	C	C	C	20	17
HC95	SA Black	+	12	18	25	26	C	C	T	C	C	C	20	17
HC144-1	SA Black	+	12	14	25	14	C	C	T	C	C	C	20	25
HC144 -2	SA Black	+	12	17	25	25	C	C	T	C	C	C	20	24
HC165	SA Black	+	12	17	25	23	C	C	T	T	C	C	20	21
HC168	SA Black	+	12	16	25	21	C	C	T	T	C	C	20	23
HC175	SA Black	+	12	11	25	20	C	C	T	T	C	C	24	26
HC188	SA Black	+	12	16	25	20	C	C	T	C	C	C	20	23
HC201	SA Black	+	12	16	25	25	C	C	T	C	C	C	20	28
HC236	SA Black	+	12	18	25	20	C	C	T	T	C	T	20	24
HC254	SA Black	+	12	21	25	23	C	C	T	T	C	T	20	20
HC224	SA Black	+	14	21	25	20	C	C	T	C	C	C	20	20
HC233	SA Black	+	20	21	25	25	C	C	T	T	C	T	20	21
HC178-1	SA Mixed Ancestry	+	12	16	25	21	C	C	T	T	C	T	20	24
HC178-2	SA Mixed Ancestry	+	12	16	25	24	C	C	T	C	C	C	20	25
HC178-3	SA Mixed Ancestry	+	12	17	25	23	C	C	T	T	C	T	20	20
HC191	SA Mixed Ancestry	+	12	23	25	23	C	C	T	T	C	C	20	22
HC193	SA Mixed Ancestry	+	12	21	25	21	C	C	T	C	C	C	23	28
55-593	African American	+	12	20	25	26	C	C	T	T	C	C	22	24
870-4376	African American	+	12	11	26	15	NR	NR	T	T	C	C	22	24
870-4934	African American	+	12	18	26	23	NR	NR	T	T	NR	NR	22	24
870-4935	African American	+	12	11	26	13	C	A	T	T	NR	NR	22	24
870-5011	African American	+	12	18	26	23	C	C	T	T	C	C	22	24
870-5504	African American	-	12	16	25	25	A	C	T	T	NR	NR	22	24
870-5790	African American	+	12	11	26	23	C	A	T	T	NR	NR	22	24
870-5792	African American	+	12	11	26	23	C	A	T	T	NR	NR	22	24
870-5793	African American	+	12	11	26	23	C	A	T	T	C	C	22	24
870-5798	African American	+	12	12	26	21	C	C	T	C	C	C	22	23
870-5800	African American	-	16	24	21	25	A	C	NR	NR	NR	NR	23	26
870-5802	African American	+	12	11	26	23	C	A	NR	NR	NR	NR	22	24
870-5804	African American	+	12	16	26	24	C	A	T	T	NR	NR	22	24
870-5856	African American	-	16	24	21	25	C	C	T	T	C	C	23	26

SA, South African
NR, No result

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Individual	Origin	<i>JPH3</i> expansion	Marker											
			D16S3074		D16S3048		(SNP1) rs2544638		(SNP2) rs3849254		(SNP3) rs918368		D16S413	
			Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2
1760-6253	African American	+	12	16	26	23	NR	NR	T	T	C	T	20	22
1465-5619	African American	+	12	16	25	23	C	C	T	C	C	C	21	21
1465-5999	African American	+	12	22	25	26	C	C	T	C	C	C	21	27
1856-6875	African American	+	12	14	25	22	C	C	T	C	C	C	23	23
1848-6859	African American	+	12	11	25	21	C	C	T	C	C	C	23	25
1827-6413	African American	+	12	17	25	26	C	C	T	T	NR	NR	22	22
1881-6917	African American	+	12	22	26	24	C	C	T	T	C	C	22	23
1705-6170	Other ¹	+	16	22	26	23	C	C	T	T	V	V	22	24
1705-6219	Other ¹	+	16	21	26	13	NR	NR	T	T	C	C	22	27
1769-6286	Mexican	+	16	21	25	21	C	C	T	T	C	C	23	21
1769-6264	Mexican	+	16	21	25	25	C	C	NR	NR	C	C	23	20
1769-6265	Mexican	-	16	23	21	24	C	C	T	T	C	C	22	23
1769-6266	Mexican	-	21	23	23	25	C	C	T	C	C	C	21	23
1769-6267	Mexican	-	16	21	21	25	T	T	T	T	C	C	22	23
1769-6268	Mexican	-	16	24	21	23	C	C	T	T	C	C	21	23
1769-6287	Mexican	-	16	23	19	23	NR	NR	T	T	C	T	22	24
1886-6927	Mexican	+	12	21	25	21	C	C	T	T	C	C	21	23
2289-001	Moroccan	+	11	23	26	21	C	T	T	T	T	T	16	23
571-005	French West Indian	+	22	17	21	21	C	T	T	T	C	C	20	22
571-002	French West Indian	-	12	17	20	21	C	T	T	T	C	C	21	22

¹European American Indian African

NR, No result

Supplementary Table 2: Genotyping Data for 3 SNP and 3 microsatellite markers at the *JPH3* locus in South African Black unaffected individuals

Individuals	Marker											
	D16S3074		D16S3048		(SNP1) rs2544638		(SNP2) rs3849254		(SNP3) rs918368		D16S413	
	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2	Allele 1	Allele 2
1	11	16	23	26	C	C	T	T	T	T	21	29
2	12	15	24	24	C	C	C	T	C	C	17	24
3	12	14	23	26	C	C	C	T	T	C	22	24
4	14	21	20	23	C	C	C	T	T	C	16	23
5	16	16	24	25	C	C	C	T	T	C	22	25
6	14	21	20	21	C	C	T	T	T	C	16	21
7	20	22	22	22	C	C	C	T	T	T	21	22
8	12	16	13	23	C	C	C	T	T	C	25	34
9	15	16	20	26	C	C	C	C	C	C	20	25
10	16	20	21	23	C	C	C	C	T	C	24	26
11	12	16	22	25	C	C	T	T	C	C	23	24
12	19	20	21	25	C	C	T	T	C	C	22	24
13	NR	NR	13	21	C	C	C	T	C	C	10	24
14	15	20	20	25	C	C	T	T	T	T	20	24
15	17	20	17	25	C	C	T	T	T	C	24	24
16	11	18	20	23	C	A	C	T	C	C	16	23
17	11	21	16	26	C	C	C	T	C	C	25	27
18	11	11	21	23	C	C	C	T	T	C	24	25
19	16	16	19	25	C	C	T	T	T	T	18	21
20	16	17	20	24	C	C	C	C	C	C	23	24
21	16	17	20	21	C	C	C	T	C	C	20	24
22	16	16	20	24	C	C	C	T	C	C	21	23
23	15	16	25	26	C	C	NR	NR	T	C	24	26
24	17	20	21	21	C	A	C	T	C	C	20	22
25	14	16	26	27	C	C	C	C	C	C	17	21
26	16	17	20	21	NR	NR	NR	NR	NR	NR	23	24
27	11	17	25	25	NR	NR	NR	NR	NR	NR	25	27
28	11	16	25	25	NR	NR	NR	NR	NR	NR	26	28
29	15	16	19	24	NR	NR	NR	NR	NR	NR	22	22
30	17	18	21	23	NR	NR	NR	NR	NR	NR	10	24
31	12	16	13	19	NR	NR	NR	NR	NR	NR	21	21
32	16	20	21	25	NR	NR	NR	NR	NR	NR	21	26

NR, No result