

Association Studies and Direct DNA Sequencing Implicate Genetic Susceptibility Loci in the Etiology of Nonsyndromic Orofacial Clefts in Sub-Saharan African Populations

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Appendix Table 1: List of 48 SNPs that were genotyped

Chromosome	coordinate	SNP	Probable gene/loci	Alleles/ Variation	NA18856 Genotype in 1000 Genomes	NA18855 Genotype in 1000 Genomes	Average Call Rate (%)	Reference study	Study population
1	11854476	rs1801131	<i>MTHFR</i>	T>G	T/T	T/T	99.4	Boyles et al.2008	Europeans
1	11856378	rs1801133	<i>MTHFR</i>	G>A	G/A	G/G	99.7	Boyles et al.2008	Europeans
1	18956458	rs766325	<i>PAX7</i>	A>G	A/A	A/A	99.6	Beaty et al. 2010	Europeans and Asians
1	18979874	rs742071	<i>PAX7</i>	G>T	T/G	G/G	99.4	Beaty et al. 2010	Europeans and Asians
1	94553438	rs560426	<i>ABCA4</i>	T>C	C/T	T/C	99.5	Beaty et al. 2010	Europeans and Asians
1	94570016	rs481931	<i>ABCA4</i>	G>T	G/G	No data	99.5	Beaty et al. 2010	Europeans and Asians
1	94575056	rs4147811	<i>ABCA4</i>	C>T	C/C	No data	99.3	Beaty et al. 2010	Europeans and Asians
1	94650805	rs138751793	<i>ARHGAP29</i>	T>C	T/T	No data	99.3	Present Study	Africans
1	108699730	rs6677101	<i>SLC25A24</i>	T>G	G/T	G/G	99.1	Butler et al. 2015	Europeans
1	209977111	rs861020	<i>IRF6</i>	G>A	G/G	G/G	99.5	Rojas-Martinez et al.2010	Europeans
1	209979529	rs34743335	<i>IRF6</i>	A>T	A/A	No data	97.9	Pegelow et al. 2008	Europeans
1	209989270	rs642961	<i>IRF6</i>	G>A	G/G	G/G	99.5	Rahimov et al. 2008	Europeans and Asians

1	210174417	rs11119388	<i>SYT14</i>	A>G	A/A	A/A	99.6	Leslie et al. 2014	Europeans and Asians
2	43540125	rs7590268	<i>THADA</i>	T>G	T/T	T/T	99.5	Mangold et al. 2010	Europeans
2	71674476	rs4332945	<i>DYSF</i>	T>G	T/T	T/T	99.3	Brayton et al. 2009	Mouse screen
2	71780215	rs2303596	<i>DYSF</i>	C>T	C/T	C/C	99.2	Brayton et al. 2009	Mouse screen
2	71866842	rs227782	<i>DYSF</i>	A>G	A/G	A/G	99.3	Brayton et al. 2009	Mouse screen
4	4865146	rs12532	<i>MSX1</i>	A>G	G/A	A/G	99.4	Suzuki et al. 2004	Asians
4	4864991	rs115200552	<i>MSX1</i>	G>C	G/G	No data	99.5	Present study	Africans
6	93506409	rs2674394	Gene Desert	A>C	C/C	C/C	99.4	Ludwig et al. 2012	Europeans and Asians
6	158885758	rs651333	<i>TULP4</i>	C>T	C/T	T/T	99.3	Ludwig et al. 2012	Europeans and Asians
8	27389542	rs6558002	<i>EPHX2</i>	C>T	C/C	C/C	99.1	Ludwig et al. 2012	Europeans and Asians
8	129946154	rs987525	8q24	A>C	A/A	C/C	99.4	Birnbaum et al. 2009	Europeans
9	100612270	rs894673	<i>FOXE1</i>	A>T	T/T	T/T	99.3	Moreno et al. 2009	Europeans, Asians and Hispanics
9	100614140	rs3758249	<i>FOXE1</i>	T>C	C/C	C/C	99.4	Moreno et al. 2009	Europeans, Asians and Hispanics
10	118827560	rs7078160	<i>VAX1</i>	G>A	A/G	A/G	99.2	Beaty et al. 2010	Europeans and Asians
10	118834991	rs4752028	<i>VAX1</i>	T>C	C/C	C/T	99.6	Beaty et al. 2010	Europeans and Asians
12	43819298	rs10785430	<i>ADAMTS20</i>	A>G	A/A	A/A	99.5	Wolf et al. 2015	Hispanics
13	80668874	rs9574565	<i>SPRY2</i>	T>C	C/T	T/T	99.4	Ludwig et al. 2012	Europeans and Asians
13	80692811	rs8001641	<i>SPRY2</i>	G>A	A/G	G/G	99.5	Ludwig et al. 2012	Europeans and Asians
13	92003297	rs375489721	<i>MIR17HG</i>	T>C	T/T	No data	99.1	Amendt et al. unpublished	Mouse screen

13	92003356	rs185831554	<i>MIR17HG</i>	T>G	T/T	No data	99.3	Amendt et al. unpublished	Mouse screen
14	54417522	rs17563	<i>BMP4</i>	A>G	A/A	G/A	99.5	Chen et al. 2008	Asians
15	33050423	rs1258763	<i>GREM1</i>	C>T	C/C	C/C	99.3	Ludwig et al. 2012	Europeans and Asians
16	3980445	rs8049367	<i>ADCY9</i>	C>T	C/T	C/T	99.4	Sun et al. 2015	Asians
16	68771034	rs16260	<i>CDH1</i>	C>A	C/A	A/C	99.2	Song et al. 2011	Asians
16	68790394	rs11642413	<i>CDH1</i>	G>A	G/G	A/G	99.5	Song et al. 2011	Asians
16	84872051	rs1546124	<i>CRISPLD2</i>	C>G	C/C	C/G	99.5	Chiquet et al. 2007	Hispanics
16	84941329	rs4783099	<i>CRISPLD2</i>	C>T	C/T	C/C	99.3	Chiquet et al. 2007	Hispanics
17	8956285	rs8069536	<i>NTN1</i>	G>T	G/G	G/T	99.4	Beaty et al. 2010	Europeans and Asians
17	8965551	rs8081823	<i>NTN1</i>	G>A	A/G	G/G	99.4	Beaty et al. 2010	Europeans and Asians
17	54615617	rs17760296	<i>NOG1</i>	T>G	T/T	T/T	99.5	Mangold et al. 2010	Europeans
17	54773238	rs227731	<i>NOG1</i>	G>T	T/G	T/G	99.3	Mangold et al. 2010	Europeans
17	63528123	rs7224837	<i>AXIN2</i>	A>G	A/G	A/A	99.3	Letra et al. 2012	Europeans and Asians
17	63549488	rs3923086	<i>AXIN2</i>	A>C	A/A	A/A	99.7	Letra et al. 2012	Europeans and Asians
20	39268516	rs17820943	<i>MAFB</i>	C>T	T/C	C/C	99.6	Beaty et al. 2010	Europeans and Asians
20	39269074	rs13041247	<i>MAFB</i>	T>C	C/T	T/T	99.5	Beaty et al. 2010	Europeans and Asians
20	39270816	rs11696257	<i>MAFB</i>	C>T	T/C	C/C	99.3	Beaty et al. 2010	Europeans and Asians

Note: Except the studies designated as “present study” and Moreno et al. 2009, these loci were largely associated with NSCL/P in the study populations.