

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 2: Case-control analyses for Ethiopia

Part A: Case-control analyses for NSCL/P and NSCPO for Ethiopia							
SNP	Probable gene/loci	NSCL/P			NSCPO		
		<i>p</i>	OR	95% CI	<i>p</i>	OR	95% CI
rs1801131	<i>MTHFR</i>	0.37	1.10	0.89 - 1.36	0.88	0.95	0.49 - 1.85
rs1801133	<i>MTHFR</i>	0.85	0.97	0.71 - 1.34	0.83	0.89	0.31 - 2.54
rs766325	<i>PAX7</i>	0.39	0.90	0.71 - 1.14	0.86	0.94	0.46 - 1.92
rs742071	<i>PAX7</i>	5.57E-03^a	1.33	1.09 - 1.63	0.53	0.82	0.44 - 1.53
rs560426	<i>ABCA4</i>	0.95	0.99	0.81 - 1.22	0.23	1.46	0.79 - 2.71
rs481931	<i>ABCA4</i>	0.75	0.95	0.68 - 1.32	0.03^a	0.00	0.00 - NA
rs4147811	<i>ABCA4</i>	0.69	0.94	0.67 - 1.30	0.03^a	0.00	0.00 - NA
rs138751793	<i>ARHGAP29</i>	0.62	0.59	0.07 - 4.88	0.05	6.42	0.73 - 56.15
rs6677101	<i>SLC25A24</i>	0.30	0.89	0.72 - 1.11	0.52	1.23	0.66 - 2.29
rs861020	<i>IRF6</i>	0.11	1.21	0.96 - 1.52	0.13	1.66	0.86 - 3.19
rs34743335	<i>IRF6</i>	0.27	0.78	0.51 - 1.21	0.52	0.63	0.15 - 2.63
rs642961	<i>IRF6</i>	0.02^a	1.44	1.07 - 1.94	0.22	1.68	0.73 - 3.84
rs7590268	<i>THADA</i>	0.47	0.92	0.73 - 1.16	0.70	1.14	0.58 - 2.26
rs4332945	<i>DYSF</i>	0.45	0.92	0.74 - 1.14	0.77	0.91	0.47 - 1.74
rs2303596	<i>DYSF</i>	2.31E-03^a	0.69	0.54 - 0.87	0.10	0.51	0.22 - 1.15
rs227782	<i>DYSF</i>	0.10	0.84	0.68 - 1.04	0.09	0.55	0.27 - 1.10
rs115200552	<i>MSX1</i>	0.40	1.46	0.60 - 3.55	0.33	2.66	0.34 - 20.96
rs12532	<i>MSX1</i>	0.59	1.06	0.86 - 1.31	0.22	0.65	0.32 - 1.31
rs2674394	Gene Desert	0.69	0.95	0.74 - 1.23	0.73	1.14	0.54 - 2.41
rs651333	<i>TULP4</i>	0.52	0.94	0.76 - 1.15	0.70	0.88	0.47 - 1.67
rs6558002	<i>EPHX2</i>	0.44	0.92	0.76 - 1.13	0.44	0.78	0.42 - 1.46
rs987525	8q24	7.82E-04^a	1.41	1.15 - 1.73	0.20	1.50	0.81 - 2.78
rs894673	<i>FOXE1</i>	0.47	0.93	0.75 - 1.15	0.53	1.23	0.65 - 2.29
rs3758249	<i>FOXE1</i>	0.52	0.93	0.75 - 1.16	0.52	1.23	0.66 - 2.30
rs7078160	<i>VAX1</i>	0.48	1.10	0.85 - 1.41	0.56	0.77	0.32 - 1.86

rs4752028	VAX1	0.90	0.99	0.81 - 1.21	0.50	0.80	0.42 - 1.52
rs10785430	ADAMTS20	0.83	1.02	0.82 - 1.27	0.69	1.14	0.59 - 2.19
rs9574565	SPRY2	0.87	0.98	0.80 - 1.21	0.57	0.83	0.43 - 1.59
rs8001641	SPRY2	0.21	1.15	0.92 - 1.43	0.19	0.59	0.27 - 1.30
rs17563	BMP4	0.91	0.99	0.80 - 1.22	0.75	0.90	0.46 - 1.75
rs1258763	GREM1	0.15	0.86	0.70 - 1.06	0.79	1.09	0.59 - 2.02
rs8049367	ADCY9	0.95	0.99	0.80 - 1.23	0.36	0.72	0.36 - 1.46
rs16260	CDH1	0.46	1.11	0.84 - 1.48	0.66	0.81	0.31 - 2.08
rs11642413	CDH1	0.39	1.09	0.89 - 1.34	0.84	1.07	0.58 - 1.97
rs1546124	CRISPLD2	0.44	0.92	0.75 - 1.13	0.36	0.74	0.39 - 1.41
rs4783099	CRISPLD2	0.87	0.98	0.80 - 1.21	0.17	0.62	0.32 - 1.23
rs8069536	NTN1	0.77	1.04	0.81 - 1.32	0.15	0.51	0.20 - 1.31
rs8081823	NTN1	0.20	0.87	0.69 - 1.08	0.04^a	0.41	0.17 - 0.98
rs17760296	NOG1	0.48	0.91	0.70 - 1.18	0.51	1.28	0.62 - 2.63
rs227731	NOG1	0.45	0.93	0.76 - 1.13	0.71	0.89	0.48 - 1.65
rs7224837	AXIN2	0.53	1.16	0.73 - 1.85	0.16	0.00	0.00 - NA
rs3923086	AXIN2	0.42	1.11	0.87 - 1.41	0.59	1.22	0.59 - 2.52
rs17820943	MAFB	0.06	0.81	0.65 - 1.01	0.27	0.68	0.34 - 1.36
rs13041247	MAFB	0.04^a	0.80	0.64 - 0.99	0.51	0.80	0.41 - 1.55
rs11696257	MAFB	0.04^a	0.79	0.64 - 0.99	0.51	0.80	0.41 - 1.55

Part B: Case-control analyses for NSCL/P subphenotypes for Ethiopia

SNP	Probable gene/loci	NSCL			NSCLP		
		<i>p</i>	OR	95% CI	<i>p</i>	OR	95% CI
rs1801131	MTHFR	0.37	1.15	0.85 - 1.57	0.57	1.08	0.82 - 1.42
rs1801133	MTHFR	0.42	0.81	0.49 - 1.34	0.58	1.12	0.75 - 1.66
rs766325	PAX7	0.68	0.93	0.66 - 1.31	0.56	0.91	0.68 - .123
rs742071	PAX7	7.74E-03^a	1.49	1.11 - 2.00	0.06	1.28	0.99 - 1.65
rs560426	ABCA4	0.42	0.88	0.66 - 1.19	0.57	1.08	0.83 - 1.40
rs481931	ABCA4	0.70	0.91	0.56 - 1.49	0.91	1.03	0.67 - 1.56
rs4147811	ABCA4	0.62	0.88	0.54 - 1.44	0.96	1.01	0.66 - 1.53
rs138751793	ARHGAP29	0.45	0.00	0.00 - NA	0.94	0.93	0.11 - 7.76
rs6677101	SLC25A24	0.93	0.99	0.72 - 1.34	0.30	0.87	0.66 - 1.14
rs861020	IRF6	0.07	1.35	0.98 - 1.87	0.31	1.17	0.87 - 1.57
rs34743335	IRF6	0.53	0.82	0.44 - 1.53	0.26	0.73	0.42 - 1.27
rs642961	IRF6	0.49	1.18	0.74 - 1.86	9.11E-03^a	1.61	1.12 - 2.31
rs7590268	THADA	0.17	0.78	0.54 - 1.11	0.98	1.00	0.75 - 1.34
rs4332945	DYSF	0.14	0.79	0.57 - 1.08	0.96	1.01	0.77 - 1.32
rs2303596	DYSF	4.99E-03^a	0.59	0.41 - 0.86	0.03^a	0.70	0.52 - 0.96
rs227782	DYSF	0.41	0.88	0.65 - 1.19	0.17	0.83	0.63 - 1.09
rs115200552	MSX1	0.04^a	2.81	0.99 - 7.97	0.69	0.74	0.17 - 3.26
rs12532	MSX1	0.82	1.04	0.76 - 1.41	0.58	1.08	0.83 - 1.41

rs2674394	Gene Desert	0.70	1.07	0.75 - 1.54	0.60	0.91	0.66 - 1.27
rs651333	<i>TULP4</i>	0.35	0.86	0.64 - 1.17	0.79	0.96	0.74 - 1.25
rs6558002	<i>EPHX2</i>	0.79	0.96	0.72 - 1.29	0.35	0.88	0.68 - 1.15
rs987525	8q24	0.03^a	1.38	1.03 - 1.85	1.07E-03^a	1.54	1.19 - 1.99
rs894673	<i>FOXE1</i>	0.17	0.80	0.58 - 1.10	0.99	1.00	0.77 - 1.31
rs3758249	<i>FOXE1</i>	0.12	0.78	0.56 - 1.07	0.79	1.04	0.79 - 1.36
rs7078160	<i>VAX1</i>	0.87	0.97	0.66 - 1.43	0.20	1.23	0.89 - 1.68
rs4752028	<i>VAX1</i>	0.85	0.97	0.72 - 1.31	0.99	1.00	0.77 - 1.30
rs10785430	<i>ADAMTS20</i>	0.14	1.26	0.93 - 1.72	0.36	0.87	0.66 - 1.16
rs9574565	<i>SPRY2</i>	7.05E-03^a	1.50	1.11 - 2.01	0.02^a	0.73	0.55 - 0.95
rs8001641	<i>SPRY2</i>	0.72	0.94	0.68 - 1.31	0.09	1.27	0.97 - 1.67
rs17563	<i>BMP4</i>	0.93	0.99	0.72 - 1.35	0.91	1.02	0.77 - 1.33
rs1258763	<i>GREM1</i>	0.44	0.89	0.66 - 1.20	0.15	0.82	0.63 - 1.07
rs8049367	<i>ADCY9</i>	0.69	0.94	0.68 - 1.29	0.81	1.03	0.79 - 1.36
rs16260	<i>CDH1</i>	0.83	1.05	0.69 - 1.59	0.58	1.11	0.77 - 1.59
rs11642413	<i>CDH1</i>	0.27	1.18	0.88 - 1.58	0.66	1.06	0.82 - 1.37
rs1546124	<i>CRISPLD2</i>	0.47	0.90	0.66 - 1.21	0.62	0.94	0.72 - 1.21
rs4783099	<i>CRISPLD2</i>	0.49	0.90	0.66 - 1.22	0.72	1.05	0.81 - 1.36
rs8069536	<i>NTN1</i>	0.65	0.92	0.64 - 1.33	0.28	1.18	0.87 - 1.59
rs8081823	<i>NTN1</i>	0.84	0.97	0.70 - 1.33	0.30	0.86	0.64 - 1.15
rs17760296	<i>NOG1</i>	0.42	0.85	0.58 - 1.26	0.68	0.93	0.67 - 1.30
rs227731	<i>NOG1</i>	0.19	0.82	0.61 - 1.10	0.84	0.97	0.75 - 1.26
rs7224837	<i>AXIN2</i>	0.61	1.19	0.62 - 2.29	0.54	1.20	0.67 - 2.18
rs3923086	<i>AXIN2</i>	0.79	1.05	0.73 - 1.50	0.19	1.22	0.90 - 1.66
rs17820943	<i>MAFB</i>	0.02^a	0.68	0.48 - 0.95	0.24	0.85	0.64 - 1.12
rs13041247	<i>MAFB</i>	0.02^a	0.67	0.48 - 0.93	0.22	0.84	0.64 - 1.11
rs11696257	<i>MAFB</i>	0.02^a	0.67	0.48 - 0.93	0.19	0.83	0.63 - 1.09

^aLoci that reached nominal significance