

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 3: Case-control analyses for Ghana

Part A: Case-control analyses for NSCL/P and NSCPO for Ghana							
SNP	Probable gene/loci	NSCL/P			NSCPO		
		<i>p</i>	OR	95% CI	<i>p</i>	OR	95% CI
rs1801131	<i>MTHFR</i>	0.82	1.03	0.79 - 1.35	0.40	0.81	0.51 - 1.32
rs1801133	<i>MTHFR</i>	0.94	1.01	0.72 - 1.43	0.51	0.81	0.44 - 1.49
rs766325	<i>PAX7</i>	0.82	0.97	0.75 - 1.26	0.65	0.91	0.60 - 1.38
rs742071	<i>PAX7</i>	0.84	1.02	0.85 - 1.23	0.88	0.98	0.72 - 1.33
rs560426	<i>ABCA4</i>	<b>0.03<sup>a</sup></b>	<b>1.22</b>	<b>1.01 - 1.47</b>	0.16	1.24	0.92 - 1.67
rs481931	<i>ABCA4</i>	0.67	1.07	0.78 - 1.49	0.47	0.81	0.46 - 1.43
rs4147811	<i>ABCA4</i>	0.89	1.02	0.74 - 1.42	0.64	0.88	0.50 - 1.52
rs138751793	<i>ARHGAP29</i>	0.77	1.10	0.57 - 2.12	0.68	0.77	0.23 - 2.63
rs6677101	<i>SLC25A24</i>	0.33	1.10	0.91 - 1.34	0.27	1.19	0.87 - 1.63
rs861020	<i>IRF6</i>	0.88	0.98	0.72 - 1.32	0.27	0.73	0.41 - 1.28
rs34743335	<i>IRF6</i>	0.93	1.04	0.43 - 2.50	0.52	0.52	0.07 - 4.07
rs642961	<i>IRF6</i>	0.48	0.89	0.63 - 1.24	0.13	0.60	0.30 - 1.17
rs7590268	<i>THADA</i>	0.80	1.03	0.82 - 1.30	0.26	0.79	0.52 - 1.20
rs4332945	<i>DYSF</i>	0.87	1.02	0.79 - 1.32	0.70	0.92	0.61 - 1.40
rs2303596	<i>DYSF</i>	0.99	1.00	0.80 - 1.25	0.05	1.40	1.00 - 1.98
rs227782	<i>DYSF</i>	0.75	1.03	0.85 - 1.25	0.30	1.17	0.87 - 1.59
rs115200552	<i>MSX1</i>	0.24	1.28	0.85 - 1.91	0.06	1.72	0.97 - 3.03
rs12532	<i>MSX1</i>	0.33	0.91	0.76 - 1.10	0.87	1.03	0.76 - 1.38
rs2674394	Gene Desert	0.55	1.08	0.84 - 1.38	0.75	1.07	0.71 - 1.60
rs651333	<i>TULP4</i>	0.74	0.97	0.79 - 1.18	0.14	1.27	0.93 - 1.73
rs6558002	<i>EPHX2</i>	0.52	1.08	0.86 - 1.35	0.67	0.92	0.62 - 1.36
rs987525	8q24	0.11	0.85	0.70 - 1.04	0.80	0.96	0.70 - 1.31
rs894673	<i>FOXE1</i>	0.24	0.89	0.73 - 1.08	0.56	0.91	0.66 - 1.25
rs3758249	<i>FOXE1</i>	0.30	0.90	0.74 - 1.10	0.59	0.92	0.67 - 1.26
rs7078160	<i>VAX1</i>	<b>0.03<sup>a</sup></b>	<b>1.25</b>	<b>1.02 - 1.54</b>	0.59	1.10	0.78 - 1.55
rs4752028	<i>VAX1</i>	0.12	0.86	0.72 - 1.04	0.54	0.91	0.67 - 1.23
rs10785430	<i>ADAMTS20</i>	0.46	0.93	0.75 - 1.14	0.20	1.23	0.89 - 1.70

rs9574565	<i>SPRY2</i>	0.45	1.08	0.89 - 1.30	0.47	1.12	0.83 - 1.51
rs8001641	<i>SPRY2</i>	0.79	1.04	0.78 - 1.40	0.55	0.86	0.53 - 1.41
rs17563	<i>BMP4</i>	0.45	0.91	0.72 - 1.16	0.99	1.00	0.69 - 1.45
rs1258763	<i>GREM1</i>	0.63	1.05	0.87 - 1.26	0.65	0.93	0.69 - 1.26
rs8049367	<i>ADCY9</i>	0.34	1.10	0.90 - 1.34	0.48	0.89	0.64 - 1.23
rs16260	<i>CDH1</i>	0.84	1.03	0.78 - 1.37	0.46	0.84	0.52 - 1.35
rs11642413	<i>CDH1</i>	0.81	0.97	0.78 - 1.21	0.24	0.80	0.55 - 1.16
rs1546124	<i>CRISPLD2</i>	0.92	1.01	0.80 - 1.28	0.44	1.15	0.80 - 1.65
rs4783099	<i>CRISPLD2</i>	0.41	1.08	0.90 - 1.31	0.28	0.84	0.61 - 1.15
rs8069536	<i>NTN1</i>	0.70	1.04	0.86 - 1.27	0.88	0.98	0.71 - 1.34
rs8081823	<i>NTN1</i>	0.46	0.92	0.75 - 1.14	0.73	1.06	0.76 - 1.47
rs17760296	<i>NOG1</i>	0.90	1.05	0.48 - 2.28	0.07	2.66	0.90 - 7.87
rs227731	<i>NOG1</i>	0.43	1.10	0.87 - 1.38	0.25	1.23	0.86 - 1.75
rs7224837	<i>AXIN2</i>	0.80	1.04	0.75 - 1.44	0.49	0.81	0.44 - 1.48
rs3923086	<i>AXIN2</i>	0.36	1.99	0.44 - 8.92	0.33	0.00	0.00 - NA
rs17820943	<i>MAFB</i>	0.88	1.02	0.81 - 1.28	0.82	1.04	0.73 - 1.49
rs13041247	<i>MAFB</i>	0.70	1.05	0.83 - 1.32	0.58	1.11	0.77 - 1.59
rs11696257	<i>MAFB</i>	0.84	1.02	0.81 - 1.29	0.88	1.03	0.72 - 1.48

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

SNP	Probable gene/loci	NSCL			NSCLP		
		<i>p</i>	OR	95% CI	<i>p</i>	OR	95% CI
rs1801131	<i>MTHFR</i>	0.58	0.90	0.63 - 1.30	0.41	1.17	0.81 - 1.68
rs1801133	<i>MTHFR</i>	0.91	1.03	0.64 - 1.64	0.92	1.02	0.64 - 1.63
rs766325	<i>PAX7</i>	0.57	1.10	0.79 - 1.53	0.30	0.82	0.57 - 1.19
rs742071	<i>PAX7</i>	0.83	0.97	0.76 - 1.25	0.48	1.10	0.85 - 1.42
rs560426	<i>ABCA4</i>	0.38	1.12	0.87 - 1.42	<b>0.01<sup>a</sup></b>	<b>1.39</b>	<b>1.08 - 1.80</b>
rs481931	<i>ABCA4</i>	0.82	1.05	0.68 - 1.62	0.76	1.07	0.69 - 1.67
rs4147811	<i>ABCA4</i>	0.57	0.88	0.55 - 1.39	0.47	1.17	0.76 - 1.80
rs138751793	<i>ARHGAP29</i>	0.54	0.71	0.24 - 2.09	0.36	1.43	0.66 - 3.09
rs6677101	<i>SLC25A24</i>	0.98	1.00	0.77 - 1.31	0.09	1.26	0.97 - 1.65
rs861020	<i>IRF6</i>	0.73	1.07	0.73 - 1.57	0.62	0.90	0.58 - 1.38
rs34743335	<i>IRF6</i>	0.89	1.08	0.35 - 3.30	0.87	0.90	0.26 - 3.15
rs642961	<i>IRF6</i>	0.89	0.97	0.63 - 1.50	0.39	0.81	0.50 - 1.32
rs7590268	<i>THADA</i>	0.86	0.97	0.71 - 1.33	0.25	1.20	0.88 - 1.64
rs4332945	<i>DYSF</i>	0.88	1.03	0.73 - 1.43	0.82	1.04	0.73 - 1.48
rs2303596	<i>DYSF</i>	0.46	1.12	0.83 - 1.50	0.69	0.94	0.68 - 1.29
rs227782	<i>DYSF</i>	0.55	0.93	0.72 - 1.19	0.20	1.18	0.91 - 1.53
rs115200552	<i>MSX1</i>	0.33	1.31	0.76 - 2.27	0.39	1.27	0.73 - 2.18
rs12532	<i>MSX1</i>	0.68	0.95	0.74 - 1.21	0.16	0.83	0.64 - 1.08
rs2674394	Gene Desert	0.08	1.32	0.97 - 1.80	0.46	0.87	0.60 - 1.26
rs651333	<i>TULP4</i>	0.72	1.05	0.81 - 1.36	0.44	0.90	0.68 - 1.18

rs6558002	<i>EPHX2</i>	0.78	0.96	0.70 - 1.30	0.14	1.25	0.93 - 1.69
rs987525	8q24	0.06	0.78	0.60 - 1.02	0.56	0.92	0.70 - 1.21
rs894673	<i>FOXE1</i>	0.43	0.90	0.70 - 1.17	0.31	0.87	0.66 - 1.14
rs3758249	<i>FOXE1</i>	0.46	0.91	0.70 - 1.17	0.41	0.89	0.68 - 1.17
rs7078160	<i>VAX1</i>	<b>0.04<sup>a</sup></b>	<b>1.32</b>	<b>1.02- 1.72</b>	0.17	1.22	0.92 - 1.63
rs4752028	<i>VAX1</i>	0.11	0.82	0.64 - 1.05	0.27	0.87	0.67 - 1.12
rs10785430	<i>ADAMTS20</i>	0.48	0.91	0.69 - 1.19	0.83	0.97	0.73 - 1.29
rs9574565	<i>SPRY2</i>	0.19	1.18	0.92 - 1.50	0.70	0.95	0.73 - 1.24
rs8001641	<i>SPRY2</i>	0.65	1.09	0.75 - 1.59	0.75	0.93	0.62 - 1.41
rs17563	<i>BMP4</i>	0.49	0.89	0.65 - 1.23	0.61	0.92	0.66 - 1.28
rs1258763	<i>GREM1</i>	0.45	1.10	0.86 - 1.40	0.65	1.06	0.82 - 1.37
rs8049367	<i>ADCY9</i>	0.56	1.08	0.83 - 1.40	0.49	1.10	0.84 - 1.44
rs16260	<i>CDH1</i>	0.10	0.70	0.46 - 1.07	0.12	1.33	0.93 - 1.91
rs11642413	<i>CDH1</i>	0.85	0.97	0.73 - 1.30	1.00	1.00	0.74 - 1.35
rs1546124	<i>CRISPLD2</i>	0.49	0.89	0.65 - 1.23	0.30	1.18	0.86 - 1.61
rs4783099	<i>CRISPLD2</i>	0.78	1.04	0.81 - 1.33	0.17	1.20	0.92 - 1.56
rs8069536	<i>NTN1</i>	0.66	0.94	0.72 - 1.23	0.31	1.15	0.88 - 1.50
rs8081823	<i>NTN1</i>	0.80	0.96	0.73 - 1.27	0.33	0.86	0.64 - 1.16
rs17760296	<i>NOG1</i>	0.71	1.21	0.44 - 3.32	0.66	1.29	0.42 - 3.99
rs227731	<i>NOG1</i>	0.61	1.08	0.80 - 1.46	0.53	1.11	0.81 - 1.52
rs7224837	<i>AXIN2</i>	0.62	1.11	0.73 - 1.69	0.80	1.06	0.67 - 1.68
rs3923086	<i>AXIN2</i>	0.15	2.86	0.64 - 12.83	0.40	0.00	0.00 - NA
rs17820943	<i>MAFB</i>	0.93	1.01	0.75 - 1.36	0.72	0.94	0.68 - 1.30
rs13041247	<i>MAFB</i>	0.80	1.04	0.77 - 1.40	0.99	1.00	0.72 - 1.38
rs11696257	<i>MAFB</i>	0.87	1.02	0.76 - 1.38	0.76	0.95	0.69 - 1.31

<sup>a</sup>Loci that reached nominal significance