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Genome-Wide Scan for Parent-of-Origin Effects in a sub-Saharan African Cohort With Nonsyndromic Cleft Lip and/or Cleft Palate (CL/P)

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Table 1: Subphenotype distribution of nonsyndromic orofacial cleft case-parent trios

Cleft subphenotype	Total number of cases
Cleft lip only (CL)	64
Cleft Palate only (CPO)	36
Cleft lip and palate (CLP)	72
Unknown cleft subphenotype	2
Total number of case-parent trios	174

Table 2: Loci with hints of possible parent-of-origin effects in nonsyndromic orofacial clefts.

Chr.	SNP ID	Coordinate (GRCh37/hg19)	Alleles		CpG island?	Probable tagging gene	P_PAT	P_MAT	Z_POO	P_POO
			Effect* allele	other allele						
1	rs6702422	211804675	G	A	N	<i>LINC01693</i>	0.06	3.49E-03	3.32	8.91E-04
1	rs16824627	1962633	G	A	N	<i>GABRD</i>	1.25E-03	0.12	3.29	1.01E-03
1	rs3795279	1959121	G	A	N	<i>GABRD</i>	2.07E-03	0.10	3.25	1.16E-03
1	rs17129327	67143976	A	G	N	<i>SGIP1</i>	1.75E-03	0.12	-3.24	1.19E-03
1	rs10494431	165079632	G	A	N	Intergenic	0.09	3.76E-03	-3.22	1.26E-03
1	rs12069077	210838533	A	G	N	<i>HHAT</i>	8.56E-04	0.17	-3.21	1.33E-03
1	rs17130976	69928349	G	A	Y	Intergenic	0.09	2.86E-03	3.13	1.77E-03
2	rs57792200	237125730	G	A	N	<i>ASB18</i>	6.58E-04	0.13	-3.48	5.12E-04
2	JHU	213801332	C	A	N	Intergenic	0.35	9.45E-05	3.39	6.93E-04
2	rs1018342	73471653	A	C	N	<i>CCT7</i>	1.18E-03	0.10	-3.38	7.38E-04
2	JHU	73474705	A	T	N	<i>CCT7</i>	1.18E-03	0.10	-3.38	7.38E-04
2	rs10181106	164960360	A	C	N	Intergenic	0.05	5.58E-03	-3.30	9.59E-04
2	rs2175793	101305327	G	A	N	Intergenic	0.14	1.43E-03	-3.22	1.26E-03
2	rs4233622	236535395	G	A	N	<i>AGAP1</i>	0.06	8.65E-03	3.10	1.91E-03
2	JHU	25510603	C	G	Y	<i>DNMT3A</i>	0.11	3.55E-03	3.10	1.94E-03
2	rs6718847	31293816	C	A	N	<i>GALNT14</i>	0.19	1.91E-03	-3.10	1.95E-03
2	rs1718709	34112561	A	G	N	<i>LINC01317</i>	0.06	7.96E-03	3.09	2.01E-03
2	kpg	2119049	G	A	Y	<i>MYT1L</i>	3.65E-03	0.10	-3.09	2.03E-03
2	rs72869496	139465387	A	G	N	<i>NXP2</i>	0.23	7.96E-04	3.08	2.10E-03
3	rs79443013	143481878	T	A	N	<i>SLC9A9</i>	1.70E-03	0.06	-3.36	7.78E-04
3	JHU	7083691	A	G	N	<i>GRM7</i>	0.11	1.32E-03	-3.19	1.41E-03
3	JHU	144825424	A	G	N	Intergenic	0.06	2.25E-03	-3.18	1.46E-03
3	JHU	83928247	G	A	N	Intergenic	0.09	1.60E-03	3.16	1.58E-03
3	rs62280570	133430137	A	G	N	<i>TF</i>	0.27	5.04E-04	-3.09	1.99E-03
4	rs17000446	76250220	A	C	N	<i>LOC441025</i>	0.33	1.61E-04	-3.34	8.47E-04
4	JHU	115282539	A	G	Y	Intergenic	0.72	2.78E-05	3.32	9.15E-04

4	JHU	190264818	A	G	N	Intergenic	0.05	6.17E-03	-3.21	1.32E-03
4	JHU	5990541	A	G	N	cDNA FLJ46481	4.68E-03	0.06	-3.16	1.60E-03
4	rs28866526	61371592	A	G	N	Intergenic	0.13	2.65E-03	3.13	1.73E-03
5	rs79719856	178749572	A	G	N	<i>ADAMTS2</i>	2.28E-03	0.06	3.41	6.45E-04
5	rs16884635	10394801	G	A	N	<i>MARCH6</i>	0.06	5.01E-03	-3.29	9.93E-04
5	JHU	10419739	A	T	N	<i>MARCH6</i>	0.1	4.18E-03	-3.15	1.63E-03
5	JHU	175277598	G	A	N	<i>CPLX2</i>	6.07E-04	0.33	-3.14	1.70E-03
5	rs10058073	40377489	G	A	N	Intergenic	0.57	3.04E-05	3.11	1.89E-03
5	JHU	178208461	A	T	N	<i>AACSP1</i>	0.11	3.55E-03	3.10	1.94E-03
5	rs11679120 3	91708452	C	A	N	cDNA FLJ31923	3.51E-03	0.08	3.07	2.11E-03
5	rs11266402 7	163013919	G	A	N	Intergenic	1.46E-03	0.21	-3.07	2.15E-03
6	rs218291	108467024	G	A	N	<i>OSTM1-AS1</i>	0.12	1.75E-03	-3.28	1.02E-03
6	rs9380150	1392372	G	A	N	<i>ZNRD1-AS1</i>	0.06	5.19E-03	-3.22	1.27E-03
6	JHU	94144602	C	A	N	<i>EPHA7</i>	8.30E-04	0.15	3.22	1.28E-03
6	JHU	108855656	G	A	N	<i>FOXO3</i>	1.01E-03	0.22	3.18	1.48E-03
6	rs12663022	150442936	G	A	N	<i>PPP1R14C</i>	0.11	3.08E-03	3.14	1.68E-03
6	rs4711206	1285775	A	G	N	<i>ZNRD1-AS1</i>	0.06	9.44E-03	-3.09	2.02E-03
6	1kg	30123573	A	C	N	<i>TRIM10</i>	0.06	9.44E-03	-3.09	2.02E-03
6	1kg	30148063	A	G	N	<i>TRIM26/TRIM15/ TRIM10/ TRIM40</i>	0.06	9.44E-03	-3.09	2.02E-03
6	JHU	14394218	G	A	Y	Intergenic	0.09	5.82E-03	3.06	2.18E-03
6	JHU	29798793	G	A	N	<i>HLA-G</i>	0.06	0.01	-3.06	2.19E-03
6	JHU	29713369	A	G	Y	<i>HLA-F-AS1</i>	0.06	0.01	-3.06	2.23E-03
7	rs11983494	21614809	A	G	N	<i>DNAH11</i>	0.13	3.01E-03	3.16	1.59E-03
7	rs79723883	151829	G	A	N	LOC100507642	0.08	2.97E-03	-3.15	1.63E-03
7	rs11300564 0	33614219	A	G	N	<i>BBS9</i>	0.06	5.91E-03	3.10	1.93E-03
8	rs7824406	18517026	A	G	N	<i>PSD3</i>	4.15E-04	0.19	-3.31	9.49E-04
8	rs11536362	1090022	G	A	N	<i>DLGAP2</i>	9.67E-04	0.07	-3.27	1.07E-03

	5									
8	rs7835842	6723132	C	G	N	<i>DEFB1</i>	0.06	6.17E-03	-3.18	1.46E-03
8	JHU	135638054	G	A	Y	<i>ZFAT</i>	2.57E-04	0.30	-3.13	1.74E-03
9	rs10781283	71546444	A	G	Y	<i>PIP5K1B</i>	0.14	1.50E-03	-3.22	1.28E-03
9	JHU	5525607	T	A	N	<i>PLGRKT/ PDCD1LG2</i>	1.67E-03	0.16	3.18	1.46E-03
10	JHU	120592853	G	A	N	Intergenic	0.06	5.48E-03	-3.20	1.36E-03
10	JHU	77311853	A	T	N	<i>C10orf11</i>	2.86E-03	0.09	-3.13	1.74E-03
10	rs4746315	77403461	G	A	N	<i>C10orf11</i>	0.01	0.05	-3.12	1.83E-03
10	JHU	65519379	G	A	N	Intergenic	9.02E-03	0.06	3.09	2.03E-03
11	rs79403502	113991982	A	G	N	<i>ZBTB16</i>	1.76E-03	0.14	3.11	1.87E-03
11	JHU	98372041	A	G	Y	Intergenic	0.16	1.09E-03	-3.11	1.90E-03
11	rs78024561	113980638	A	T	N	<i>ZBTB16</i>	7.89E-04	0.06	3.09	2.00E-03
11	rs77307338	74045283	A	T	N	<i>PGM2L1</i>	0.07	6.07E-03	-3.07	2.16E-03
12	rs11660194 9	59501993	A	G	N	Intergenic	1.09E-03	0.20	-3.08	2.08E-03
12	rs3952720	95101357	G	C	N	Intergenic	0.09	7.21E-03	3.07	2.15E-03
12	JHU	2021699	T	A	N	<i>CACNA2D4</i>	0.06	0.01	3.05	2.29E-03
13	JHU	89501972	C	A	N	Intergenic	0.05	2.70E-03	3.14	1.68E-03
13	rs9285211	57605827	A	C	N	Intergenic	0.06	0.01	-3.08	2.10E-03
13	JHU	89355501	A	G	N	Intergenic	0.05	4.51E-03	3.06	2.21E-03
13	rs9536557	54433190	A	G	N	<i>LINC00558</i>	0.11	5.96E-03	-3.05	2.26E-03
14	rs1112330	48395998	C	A	N	Intergenic	0.05	5.21E-03	-3.28	1.03E-03
14	rs3759774	70828498	A	G	N	<i>SYNJ2BP/ COX16</i>	2.91E-04	0.34	-3.19	1.45E-03
14	JHU	25449226	A	C	N	<i>STXBP6</i>	0.05	6.17E-03	-3.17	1.50E-03
14	rs8022570	48393608	G	A	N	Intergenic	0.16	2.51E-03	-3.11	1.90E-03
15	rs2044292	86555762	A	G	N	Intergenic	9.74E-03	0.06	-3.14	1.69E-03
15	rs74032630	94216050	G	A	N	<i>LINC01579</i>	9.06E-03	0.08	-3.05	2.32E-03

16	rs7202071	9732549	G	A	N	<i>RP11-297M9.1/ RNA5SP404</i>	1.93E-03	0.07	-3.47	5.19E-04
16	rs61280649	11150071	C	G	N	<i>CLEC16A</i>	0.01	0.06	-3.07	2.12E-03
17	rs7215415	52713594	G	C	N	Intergenic	6.04E-04	0.11	3.44	5.84E-04
18	JHU	67912189	T	A	N	<i>SOCS6/ RTTN</i>	0.15	8.56E-04	-3.22	1.27E-03
18	rs35811982	32003207	A	G	N	Intergenic	6.84E-03	0.06	3.17	1.51E-03
18	JHU	51027086	A	G	N	<i>DCC</i>	1.36E-03	0.18	3.11	1.88E-03
20	JHU	56444297	A	T	N	<i>MIR4532</i>	9.01E-04	0.19	-3.29	1.02E-03
20	rs6057132	10055480	C	A	N	<i>ANKEF1</i>	0.11	4.97E-03	3.07	2.16E-03
21	rs73338844	16448591	G	A	N	<i>NRIP1</i>	0.09	2.50E-03	3.26	1.10E-03
21	rs459498	42795027	G	A	Y	<i>MX1</i>	1.62E-03	0.11	-3.11	1.90E-03
X	JHU	2024101	G	A	N	Intergenic	1.06E-03	0.24	3.05	2.32E-03

* implies effect allele that is over-transmitted in either the father or mother who contribute largely to the PoO effect. P-values of paternally over-transmitted alleles (P_PAT) or maternally over-transmitted allele (P_MAT) that is responsible for the parent-of-origin effects are boldened. SNP ID JHU and 1kg refer to SNPs that were identified in populations of African ancestry in various sequencing studies at John Hopkins University (JHU) and 1000 genomes phase 3 projects, respectively.

Table S1: Single nucleotide variants that did not exhibit unidirectional parent-of-origin effects

Chr.	SNP ID	Coordinate (GRCh37/hg19)	Alleles		Probable tagging gene	P PAT	P MAT	Z POO	P POO
			effect allele	other allele					
1	rs12080693	66984425	G	A	<i>SGIP1</i>	0.04	6.17E-03	-3.28	1.05E-03
2	JHU	20474703	G	A	<i>PUM2</i>	7.63E-03	7.53E-03	-3.35	8.02E-04
3	JHU	133671881	A	T	<i>SLCO2A1</i>	2.50E-03	0.02	-3.56	3.70E-04
3	rs72976837	104112931	A	G	Intergenic	0.01	0.02	3.30	9.77E-04
4	JHU	130814563	C	A	Intergenic	0.02	6.04E-03	3.39	7.04E-04
4	rs76151329	184224456	A	G	<i>WWC2</i>	5.58E-03	0.02	-3.35	7.97E-04
5	JHU	56335318	A	G	Intergenic	0.04	1.37E-03	3.71	2.07E-04
5	rs11950412	168603104	A	G	<i>SLIT3</i>	8.30E-04	0.02	-3.7	2.14E-04
5	rs75699124	24572594	G	A	<i>CDH10</i>	0.02	3.92E-03	3.61	3.04E-04
5	rs34122040	24573074	C	A	<i>CDH10</i>	2.12E-02	1.52E-02	3.29	1.00E-03
5	JHU	133264305	G	A	<i>C5orf15</i>	0.02	5.19E-03	-3.50	4.57E-04
5	JHU	2813715	G	A	Intergenic	0.02	8.64E-03	-3.49	4.89E-04
5	JHU	121989622	A	C	<i>LOC101927379</i>	1.02E-03	4.95E-02	3.46	5.31E-04
5	rs72793517	117794601	G	A	<i>HRAT56</i>	0.02	0.01	3.33	8.67E-04
6	JHU	29781010	T	A	<i>HLA-G</i>	0.03	3.92E-04	-3.91	9.14E-05
6	JHU	29735532	G	A	HLA-F/HLA-F- AS1/HCG4	0.03	7.12E-04	-3.80	1.42E-04
6	JHU	29737161	A	G	HLA-F/HLA-F- AS1/HCG4	0.03	7.12E-04	-3.80	1.42E-04
6	rs1611185	29768344	G	A	<i>HLA-G</i>	0.03	7.12E-04	-3.80	1.42E-04
6	rs2735048	1034492	A	G	HLA-F/HLA-F- AS1/HCG4	0.02	1.48E-03	-3.77	1.66E-04
6	JHU	29834198	G	A	<i>HLA-G/HLA-F</i>	0.02	5.58E-03	-3.48	5.02E-04
6	rs2517930	1045868	C	A	HLA-F/HLA-F- AS1/HCG4	1.33E-02	1.63E-02	-3.34	8.50E-04

6	JHU	29924625	C	G	<i>HLA-G/HLA-F/ HLA-G</i>	0.01	7.63E-03	-3.33	8.60E-04
6	JHU	29733837	A	T	<i>HLA-F/HLA-F- AS1/HCG4</i>	0.04	6.77E-03	-3.33	8.60E-04
7	rs38519	28037886	G	A	<i>JAZF1</i>	6.36E-03	0.03	-3.38	7.30E-04
7	JHU	127994226	G	A	<i>PRRT4</i>	8.83E-03	0.01	-3.36	7.78E-04
8	rs74811077	27833602	G	A	<i>SCARA5</i>	0.02	2.08E-03	-3.62	2.95E-04
9	rs4877980	89046282	G	A	Intergenic	6.17E-03	0.01	-3.50	4.74E-04
9	rs818882	7015330	A	C	<i>KDM4C</i>	0.01	0.02	3.34	8.25E-04
9	JHU	29107913	C	A	<i>LINGO2</i>	0.03	0.01	-3.31	9.42E-04
9	rs111870807	2181774	G	C	<i>SMARCA2</i>	0.01	0.02	3.30	9.80E-04
11	rs72880619	24624373	C	G	<i>LUZP2</i>	1.74E-03	7.63E-03	-3.67	2.44E-04
11	rs1472450	5449374	C	A	<i>HBG2/HBE1/OR51 B5</i>	0.02	0.01	3.35	7.96E-04
11	JHU	599653	A	G	<i>PHRF1</i>	9.38E-03	3.61E-03	3.34	8.43E-04
11	JHU	116044587	C	A	Intergenic	0.01	0.01	3.32	8.95E-04
12	rs12578413	3709021	G	A	<i>PRMT8/EFCAB4B</i>	0.01	0.02	3.39	7.03E-04
13	rs678844	30168388	G	A	<i>SLC7A1</i>	0.01	8.57E-05	3.79	1.49E-04
16	rs4781033	11086902	A	G	<i>CLEC16A</i>	3.19E-03	1.36E-02	-3.71	2.09E-04
16	rs2286975	11114006	A	G	<i>CLEC16A</i>	8.65E-03	7.29E-03	-3.65	2.63E-04
16	rs6498137	11042950	G	A	<i>CLEC16A</i>	2.70E-03	2.69E-02	-3.56	3.70E-04
17	rs28526123	35874074	G	C	<i>SYNRG/ DUSP14</i>	0.02	0.01	3.35	7.96E-04

Effect allele refers to the allele that is over-transmitted in the parent with the smallest p value.