Supplemental information

Trio-based GWAS identifies novel associations

and subtype-specific risk factors for cleft palate

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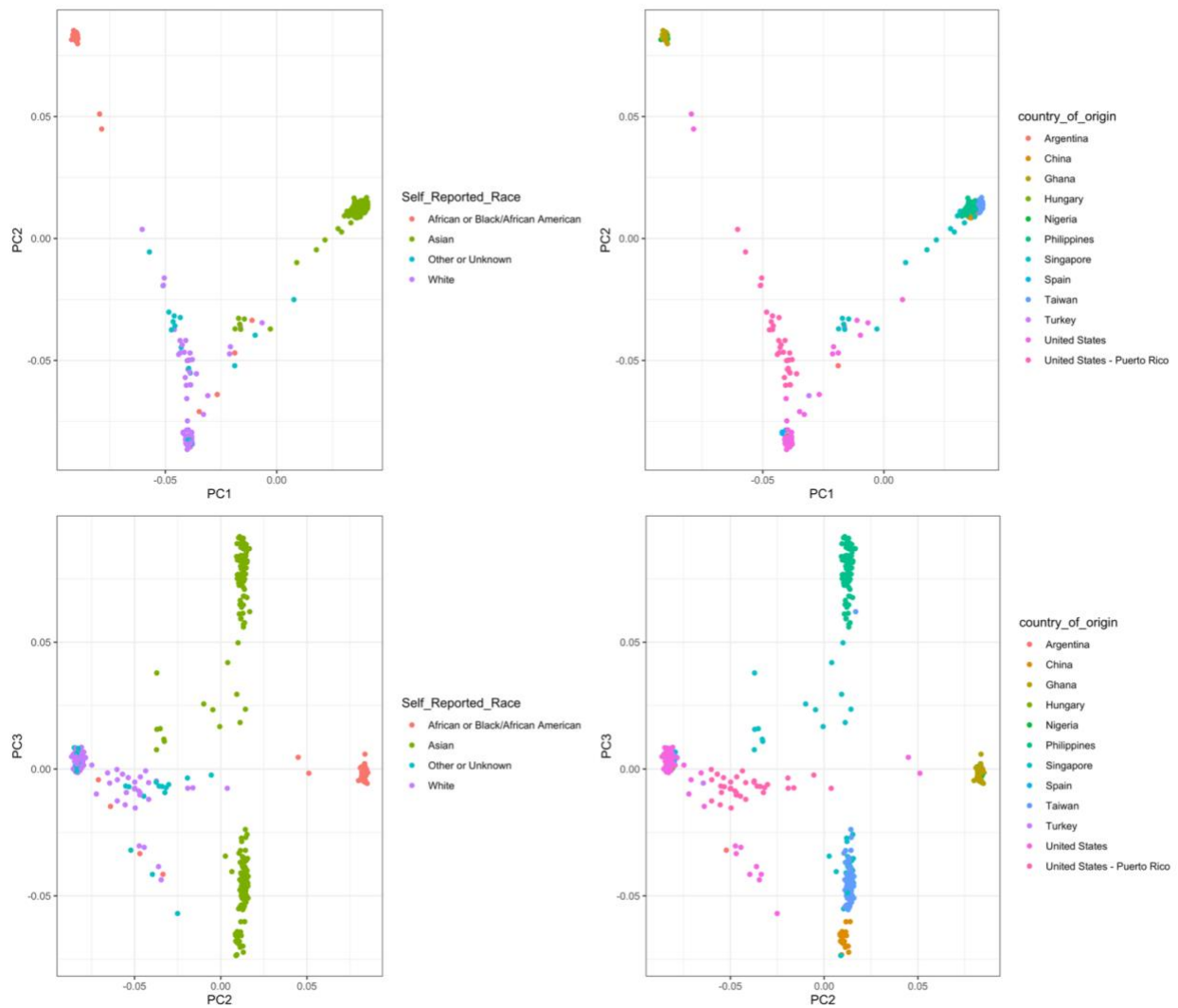Supplemental Information – Table of Contents

Figure S1: Principal components 1 vs 2 (top) and 2 vs 3 (bottom) demonstrating group separation by self-reported race and by country of origin.
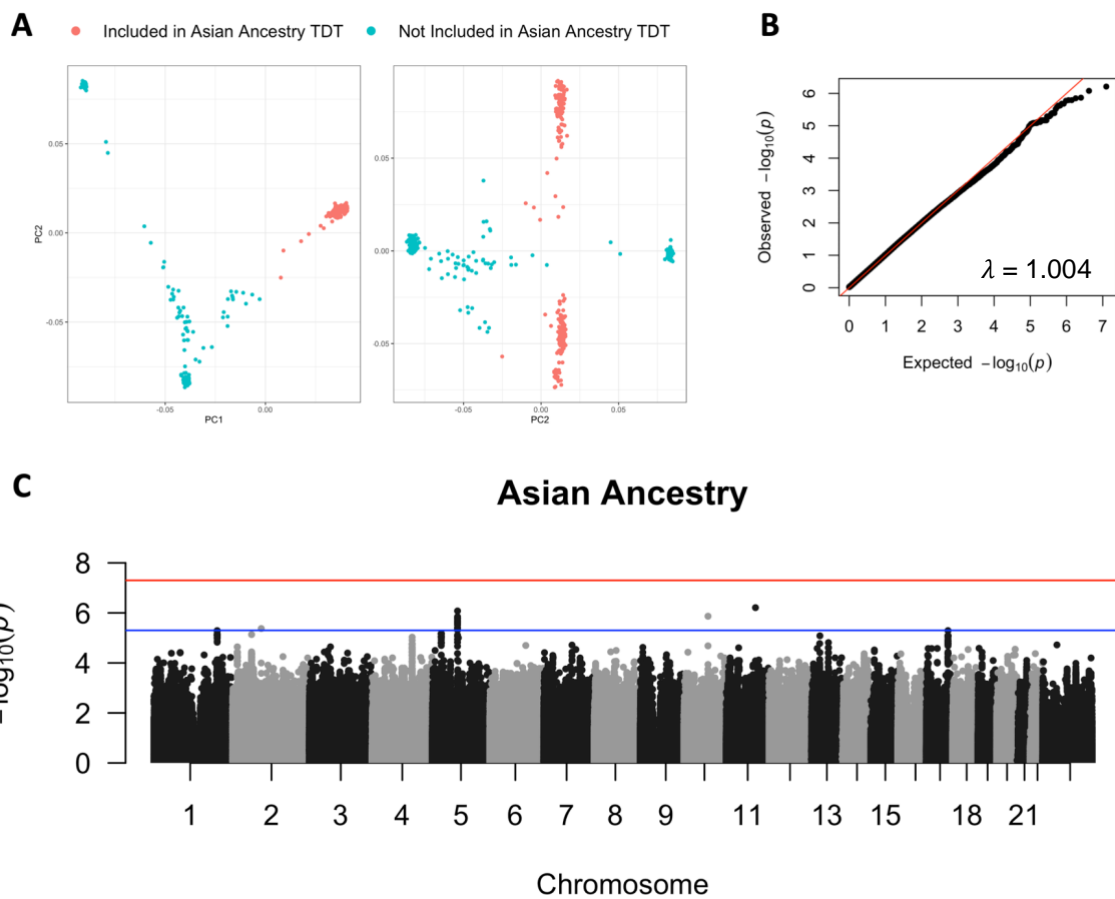
Figure S2: A) Asian ancestry as determined by PC grouping – pink dots represent probands of Asian descent (n=262). B) qqplot for Asian ancestry-specific TDT. C) Manhattan plot for proband analysis. The blue line represents the suggestive threshold (5×10^{-6}) and the red line represents genome-wide significance (5×10^{-6}).
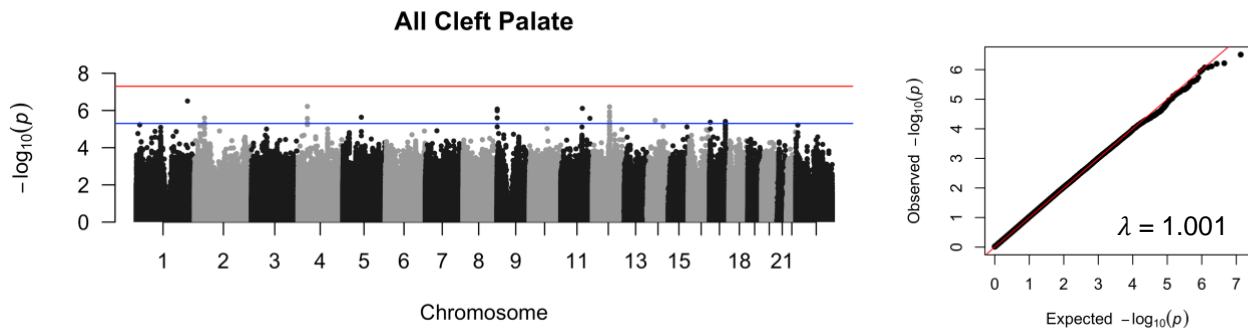
Figure S3: Manhattan plot (left) and qqplot (right) for the ACP analysis. The blue line represents the suggestive threshold (5×10^{-6}) and the red line represents genome-wide significance (5×10^{-6}).

Figure S4: Manhattan plot (left) and qqplot (right) for the CSP analysis. The blue line represents the suggestive threshold (5×10^{-6}) and the red line represents genome-wide significance (5×10^{-6}).
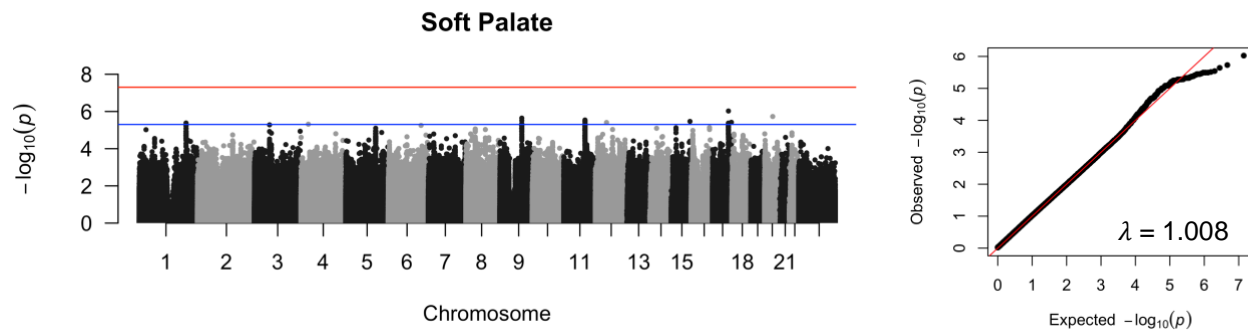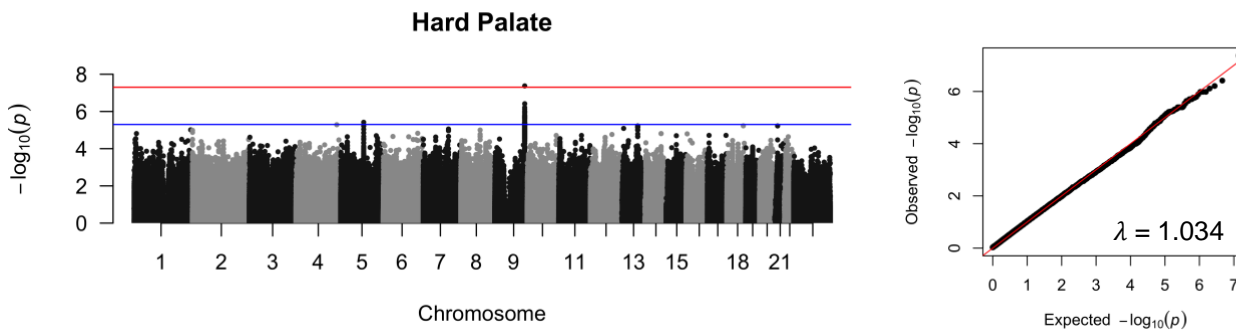

Figure S5: Manhattan plot (left) and qqplot (right) for the CHP analysis. The blue line represents the suggestive threshold (5×10^{-6}) and the red line represents genome-wide significance (5×10^{-6}).

Cleft Type	Male					
	Asian	Black/African American	White	Other	Unknown	Total Male
Both	20	6	8	1	3	38
CP - Type Unknown	20	8	10	0	9	47
Hard Palate	16	0	3	0	0	19
SMCP	2	6	6	0	0	14
Soft Palate	37	3	13	0	2	55
Total Male	95	23	40	1	14	173
Cleft Type	Female					
	Asian	Black/African American	White	Other	Unknown	Total Female
Both	31	2	8	0	3	44
CP - Type Unknown	38	24	15	0	9	86
Hard Palate	21	0	3	0	0	24
SMCP	3	7	1	0	0	11
Soft Palate	71	5	14	1	6	97
Total Female	164	38	41	1	18	262
Total Cases	259	61	81	2	32	435

Table S1: Case Description by sex and self-reported race

Gene/Direction	Sequence
Angptl2-fwd	CTGCAGAGTCTTCCAATCAG
Angptl2-rev + T7 leader	**CGATGTTAATACGACTCACTATAGGG**ATTCTGTGGAGGCTAAAGTATG

Table S2: *Angptl2* gene-specific primers

Gene/Direction	Sequence
Gapdh-fwd	AACACTGAGCATCTCCCTCACA
Gapdh-rev	GGTGGGTGCAGCGAACTTTATT
Angptl2-fwd	TCTCTGTCCCTACTGCGTTCTT
Angptl2-rev	GGCGATATTGGCAGTTGTGTCT
Runx2-fwd	GGACCCACACAGCCATGTTAAA
Runx2-rev	GGCAACGTTCCCTAACCTGAAA

Table S3: RT-qPCR primers