FaceSNPs: Identifying Face-Related SNPs from the Human Genome - Supplementary Materials

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I. Supplement A

Table I shows a sample of the genes we found through our automated literature search strategy. For each gene, we show its function as reported by the relevant literature and provide references.

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TABLE I
MANUAL LITERATURE VALIDATION OF SELECTED GENES

Chr	Genes	Function (from literature)	Region	Ref	Chr	Genes	Function (from literature)	Region	Ref
1	AJAP1	Associated with Oral Squamous Cell Carcinoma (OSCC)	mouth	[1]	13	DCLK1	Associated with macrocephaly, high forehead, hypertelorism, large nose, large and malformed ears and retrognathia	all	[2]
	DHCR24	Down-regulated in Oral Squamous Cell Carcinoma cases	mouth	[3]		KLF5	Associated with odontoblastic differentiation of dental pulp cells	mouth	[4]
2	FAM49A	Associated with nonsyndromic orofacial clefts	mouth	[5]	14	ESRRB	Essential for inner-ear development and function	ear	[6]
	HADHB	Assiciated with retinopathy	eye	[7]		SAMD4A	Associated with oral cancer	mouth	[8]
3	NISCH	Mutation associated with chronic otitis media	ear	[9]	15	TNFAIP8L3	Candidate gene for corneal astigmatism	eye	[10]
	BBX	Associated with age-related macular degeneration	eye	[11]		HERC2	Associated with Angelman Syndrome	face shape, forehead	[12]
4	EVC2	Association with microtia, Ellis-van Creveld syndrome or Weyers acrodental dysostosis	ear, mouth, face shape	[13]	16	CIITA	Associated with nasal polyposis	nose	[14]
	AFAP1	Associated with primary-open-angle glaucoma	eye	[15]		RNF40	May affect how the retina absorbs UV radiations	eye	[16]
5	SLC45A2	Associated with Oculocutaneous Albinism Type 4	eye	[17]	17	DNAH9	Associated with allergic rhinitis	nose	[18]
	COL4A3BP	Associated with several developmental disabilities	face shape	[19]		ADORA2B	May be a key regulator of tumoral progression in OSCCs	mouth	[20]
6	GMDS	Associated with primary-open-angle glaucoma	eye	[15]	18	CELF4	An important role in eye development	eye	[21]
	RIMS1	Associated with Central Areolar Choroidal Dystrophy	eye	[22]		PIEZO2	Mutation can cause a subtype of Distal Arthrogryposis Type 5	eye	[23]
7	EIF3B	Plays a role in Foot-and-mouth disease virus infection	mouth	[24]	19	OLFM2	Associated with glaucoma	eye	[25]
	CHN2	Candidate gene for cleft lip and palate	mouth	[26]		FKBP8	Associated with loss of retinal pigment epithelial cells	eye	[27]
8	ADAM3A	May be involved in the etiology of oral clefts.	mouth	[28]	20	GDF5	Associated with craniofacial dysmorphism	forehead	[29]
	CSGALNACT1	Associated with Oral Squamous Cell Carcinoma	mouth	[30]		HNF4A	Associated with genetic hearing loss	ear	[31]
9	LPAR1	Affects regulation of trabecular meshwork (TM) cell networks	eye	[32]	21	TIAM1	May contribute to the pathogenesis of extranodal natural killer (NK)/T-cell lymphoma, nasal type (ENKTCL)	nose	[33]
	PTPRD	Affects hearing functions	ear	[34]		HSPA13	Associated with OSCC occurrence	mouth	[35]
10	FRMD4A	Associated with congenital microcephaly	face shape	[36]	22	MTMR3	Possible therapeutic target for oral cancer treatment	mouth	[37]
	MPP7	Potentially associated with maxillofacial abnormalities	mouth	[38]		SYN3	Associated with larger area perifoveal ring and larger drusen volume	eye	[39]
11	BEST1	Mutations cause autosomal recessive bestrophinopathy	eye	[40]	X	CHRDL1	Associated with Oral Squamous Cell Carcinoma	mouth	[41]
	LRP5	Missense mutation cause macrocephaly, craniosynostosis	all	[42]		OCRL	Associated with Lowe syndrome	eye	[43]
12	PTPRR	Potentially associated with oral cancer	mouth	[44]	Y	AMELY	Associated with tooth enamel development	mouth	[45]
	VDR	Polymorphisms linked to initial periodontitis	mouth	[46]		UTY	Associated with nasosinusal adenocarcinoma	nose	[47]

NB: The functions are summaries from *NCBI Gene* [48]. The references our system relied on to associate the genes with the face regions are not listed due to space limitation, but are available from http://community.wvu.edu/~daadjeroh/projects/faceSNPs/FaceSNPs_BIBM2019suppl.pdf