

References:

- [1] Beaty TH, Murray JC, Marazita ML, Munger RG, Ruczinski I, Hetmanski JB, et al. A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. *Nat Genet.* 2010; 42(6): 525-9.
- [2] Beaty TH, Taub MA, Scott AF, Murray JC, Marazita ML, Schwender H, et al. Confirming genes influencing risk to cleft lip with/without cleft palate in a case-parent trio study. *Hum Genet.* 2013;132(7): 771-81.
- [3] Birnbaum S, Ludwig KU, Reutter H, Herms S, Steffens M, Rubini M, et al. Key susceptibility locus for nonsyndromic cleft lip with or without cleft palate on chromosome 8q24. *Nat Genet.* 2009; 41(4): 473-7.
- [4] Brewer C, Holloway S, Zawalnyski P, Schinzel A, Fitz-Patrick D. A chromosomal duplication map of malformations: regions of suspected haplo- and triplolethality - and tolerance of segmental aneuploidy - in humans. *Am J Hum Genet.* 1999; 64(6): 1702-1708.
- [5] Dixon MJ, Marazita ML, Beaty TH, Murray JC. Cleft lip and palate: understanding genetic and environmental influences. *Nat Rev Genet.* 2011; 12(3): 167-78.
- [6] Gong SG, Gong TW, Shum L. Identification of markers of the midface. *J Dent Res.* 2005; 84(1): 69-72.
- [7] Grant SF, Wang K, Zhang H, Glaberson W, Annaiah K, Kim CE, et al. A genome-wide association study identifies a locus for nonsyndromic cleft lip with or without cleft palate on 8q24. *J Pediatr.* 2009; 155: 909-913.

- [8] Higgins AW, Alkuraya FS, Bosco AF, Brown KK, Bruns GA, Donovan DJ, et al. Characterization of apparently balanced chromosomal rearrangements from the developmental genome anatomy project. *Am J Hum Genet.* 2008; 82(3): 712-722.
- [9] Juriloff DM, Harris MJ. Mouse genetic models of cleft lip with or without cleft palate. *Birth Defects Res A Clin Mol Teratol.* 2008; 82(2): 63-77.
- [10] Kohli SS, Kohli VS. A comprehensive review of the genetic basis of cleft lip and palate. *J Oral Maxillofac Pathol.* 2012; 16(1): 64-72.
- [11] Kondo S, Schutte BC, Richardson RJ, Bjork BC, Knight AS, Watanabe Y et al. Mutation in IRF6 cause van der Woude and popliteal pterygium syndromes. *Nat Genet.* 2002; 32(2):285-289.
- [12] Leslie EJ, Carlson JC, Shaffer JR, Feingold E, Wehby G, Laurie CA, et al. A multi-ethnic genome-wide association study identifies novel loci for non-syndromic cleft lip with or without cleft palate on 2p24.2, 17q23 and 19q13. *Hum Mol Genet.* 2016; 25(13): 2862-2872.
- [13] Ludwig KU, Ahmed ST, Bohmer AC, Sangani NB, Varghese S, Klamt J, et al. Meta-analysis Reveals Genome-Wide Significance at 15q13 for Nonsyndromic Clefting of Both the Lip and the Palate, and Functional Analyses Implicate GREM1 As a Plausible Causative Gene. *PLoS Genet.* 2016; 12(3):e1005914.
- [14] Ludwig KU, Mangold E, Herms S, Nowak S, Reutter H, Paul A, et al. Genome-wide meta-analyses of nonsyndromic cleft lip with or without cleft palate identify six new risk loci. *Nat Genet.* 2012; 44(9): 968-71.

[15] Mangold E, Ludwig KU, Birnbaum S, Baluardo C, Ferrian M, Herms S, et al. Genome-wide association study identifies two susceptibility loci for nonsyndromic cleft lip with or without cleft palate. *Nat Genet.* 2010; 42: 24-26.

[16] Mangold E, Ludwig KU, Nothen MM. Breakthroughs in the genetics of orofacial clefting. *Trends Mol Med.* 2011; 17(12): 725-33.

[17] Marazita ML, Lidral AC, Murray JC, Field LL, Maher BS, Goldstein McHenry T, et al. Genome scan, fine-mapping, and candidate gene analysis of non-syndromic cleft lip with or without cleft palate reveals phenotype-specific differences in linkage and association results. *Hum Hered.* 2009; 68(3): 151-70.

[18] Moreno LM, Mansilla MA, Bullard SA, Cooper ME, Busch TD, Machida J, et al. FOXE1 association with both isolated cleft lip with or without cleft palate, and isolated cleft palate. *Hum Mol Genet.* 2009; 18(24): 4879-96.

[19] Mukhopadhyay P, Greene RM, Zacharias W, Weinrich MC, Singh S, Young WW et al. Developmental gene expression profiling of mammalian, fetal orofacial tissue. *Birth Defects Res A Clin Mol Teratol.* 2004; 70(12): 912-926.

[20] Rahimov F, Marazita ML, Visel A, Cooper ME, Hitchler MJ, Rubini M, et al. Disruption of an AP-2alpha binding site in an IRF6 enhancer is associated with cleft lip. *Nat Genet.* 2008; 40(11): 1341-7.

[21] Sun Y, Huang Y, Yin A, Pan Y, Wang Y, Wang C, et al., Genome-wide association study identifies a new susceptibility locus for cleft lip with or without a cleft palate. *Nat Commun.* 2015; 6: 6414.

[22] Yu Y, Zuo X, He M, Gao J, Fu Y, Qin C. Genome-wide analyses of non-syndromic cleft lip with palate identify 14 novel loci and genetic heterogeneity. *Nat Commun.* 2017; 8: 14364.

[23] Zuccherro T, Cooper ME, Maher BS, Daack-Hirsch S, Nepomuceno B, Ribeiro L, et al. Interferon regulatory factor 6 (IRF6) gene variants and the risk of isolated cleft lip or palate. *Engl J Med.* 2004; 351(8):769-780.