

DAFTAR PUSTAKA

- Ahmad M, Abbas H, Haque S, Flatz G (1987) X-chromosomally inherited split-hand/split-foot anomaly in a Pakistani kindred. *Hum. Genet.* 75: 169-173.
- Amiel J, Bougeard G, Francannet C, Raclin V, Munnich A, Lyonnet S, Frebourg T (2001) TP63 gene mutation in ADULT syndrome. *Europ. J. Hum. Genet.* 9: 642-645.
- Barbieri, CE & Pietenpol, JA (2006). p63 and epithelial biology. *Exp Cell Res* 312, 695–706.
- Blattner A, Huber AR, Rothlisberger B (2010) Homozygous nonsense mutation in WNT10B and sporadic split-hand/foot malformation (SHFM) with autosomal recessive inheritance. *Am. J. Med. Genet.* 152A: 2053-2056.
- Boch J, Scholze H, Schornack S, Landgraf A, Hahn S, Kay S, Lahaye T, Nickstadt A, Bonas U (2009). "Breaking the code of DNA binding specificity of TAL-type III effectors". *Science* 326 (5959): 1509–12.
- Caselli MA. Podiatry Management January 2009 accessed on June 12th 2009 at www.podiatrym.com/cme/Jan09%20CME.pdf
- Celli J, Duijf P, Hamel BCJ, Bamshad M, Kramer B, Smits APT, Newbury-Ecob R, Hennekam RCM, van Buggenhout G, van Haeringen A, Woods CG, van Essen AJ, de Waal R, Vriend G, Haber DA, Yang A, McKeon F, Brunner HG, van Bokhoven H (1999) Heterozygous germline mutations in the p53 homolog p63 are the cause of EEC syndrome. *Cell* 99: 143-153.
- Del Campo M, Jones MC, Veraksa AN, Curry CJ, Jones KL, Mascarello JT, Ali-Kahn-Catts Z, Drumheller T, McGinnis W (1999) Monodactylous limbs and abnormal genitalia are associated with hemizyosity for the human 2q31 region that includes the HOXD cluster. *Am. J. Hum. Genet.* 65: 104-110.
- Di Iorio E, Barbaro V, Ruzza A, Ponzin D, Pellegrini G, De Luca M (2005) Isoforms of delta-N-p63 and the migration of ocular limbal cells in human corneal regeneration. *Proc. Nat. Acad. Sci.* 102: 9523-9528.
- Duijf P, van Bokhoven H, Brunner HG (2003) Pathogenesis of split-hand/split-foot -malformation. *Hum Mol Genet* 12:51-60.
- Durowaye M, Adeboye M, Yahaya-Kongoila S, Adaje A, Adesiyun O, Ernest SK, Mokuolu OA, Adegboye A (2011) Familial Ectrodactyly Syndrome in a Nigerian Child: A Case Report. *Oman Medical Journal* 26(4): 275-278.
- Faiyaz-Ul-Haque M, Zaidi SH, King LM (2005) Fine mapping of the X-linked split-hand/split-foot malformation (SHFM2) locus to a 5.1-Mb region on Xq26.3 and analysis of candidate genes. *Clin Genet* 67:93-97
- Flores ER, Tsai KY, Crowley D, Sengupta S, Yang A, McKeon F, Jacks, T (2002) p63 and p73 are required for p53-dependent apoptosis in response to DNA damage. *Nature* 416: 560-564.
- Hummel S (2013) *Ancient DNA Typing: Methods, Strategies and Applications*. New York: Springer Science & Business Media.
- Ianakeiev P, Kilpatrick MW, Toudjarska I, Basel D, Beighton P, Tsipouras P (2000) Split-Hand/Split-Foot Malformation Is Caused by Mutations in the p63 Gene on 3q27. *Am J Hum Genet* 67:59-66.

- Kang YS, Cheong HM, Moon Y, Lee IB, Kim SM, Kim HS, Jun SY, Jung SK, Kim JS, Choi JH, Cho HE, Son JS, Min NY, Lee KH (2004) Molecular Genetic Characterization of a Korean Split Hand/Split Foot Malformation (SHFM). *Moll Cells* 17(3):397-403.
- Kantaputra PN, Hamada T, Kumchai T, McGrath JA (2003) Heterozygous mutation in the SAM domain of p63 underlies Rapp-Hodgkin ectodermal dysplasia. *J. Dent. Res.* 82: 433-437.
- Kelman GJ dan Aronoff RC. (2000) Ectrodactyly-ectodermal dysplasia-clefting syndrome. *J Am Podiatr Med Assoc.* 90(9):460-464.
- Khan S, Basit S, Zimri F K, Ali N, Ali G, Ansar M dan Ahmad W (2011) A novel homozygous missense mutation in WNT10B in familial split-hand/foot malformation. *Clinical Genetics* 82: 48–55.
- Koster, MI dan Roop, DR (2004) The role of p63 in development and differentiation of the epidermis. *J Dermatol Sci* 34, 3–9.
- Leoyklang P, Siriwan P, Shotelersuk V (2006) A mutation of the p63 gene in non-syndromic cleft lip. (Letter) *J. Med. Genet.* 43: e28.
- Liu H dan Naismith JH (2008) An efficient one-step site-directed deletion, insertion, single and multiple-site plasmid mutagenesis protocol. *BMC Biotechnology.* 8:91.
- McGrath JA, Duijf PHG, Doetsch V, Irvine AD, de Waal R, Vanmolkot KRJ, Wessagowit V, Kelly A, Atherton DJ, Griffiths WAD, Orlow SJ, van Haeringen A, Aulsems MGEM, Yang A, McKeon F, Bamshad MA, Brunner HG, Hamel BCJ, van Bokhoven H (2001) Hay-Wells syndrome is caused by heterozygous missense mutations in the SAM domain of p63. *Hum. Molec. Genet.* 10: 221-229.
- Ozen RS, Baysal BE, Devlin B, Farr JE, Gorry M, Ehrlich GD (1999) Fine mapping of the split-hand/split-foot locus (SHFM3) at 10q24: evidence for anticipation and segregation distortion. *Am J Hum Genet.* 64(6):1646-1654.
- Pinette M, Garcia L, Wax JR, Cartin A, Blackstone J (2006) Familial ectrodactyly. *J Ultrasound Med.* 25:1465–7.
- Roscioli T, Taylor PJ, Bohlken A, Donald JA, Masel J, Glass IA, Buckley MF (2004) The 10q24-linked split hand/split foot syndrome (SHFM3): narrowing of the critical region and confirmation of the clinical phenotype. *Am. J. Med. Genet.* 124A: 136-141.
- Sasaki, Y., Ishida, S., Morimoto, I., Yamashita, T., Kojima, T., Kihara, C., Tanaka, T., Imai, K., Nakamura, Y. and Tokino, T. (2002) The p53 family member genes are involved in the Notch signal pathway. *J. Biol. Chem.*, 277, 719–724.
- Saunders, J.W.J. (1948) The proximo-distal sequence of the origin of the parts of the chick wing and the role of the ectoderm. *J. Exp. Zool.*, 108, 363–403.
- Seto, M.L., Nunes, M.E., MacArthur, C.A. and Cunningham, M.L. (1997) Pathogenesis of ectrodactyly in the Dactylaplasia mouse: aberrant cell death of the apical ectodermal ridge. *Teratology*, 56, 262–270.
- Shamseldin HE, Faden MA, Alashram W, Alkuraya, FS (2012) Identification of a novel DLX5 mutation in a family with autosomal recessive split hand and foot malformation. *J. Med. Genet.* 49: 16-20.



- Su X, Chakravarti D, Cho MS, Liu L, Gi YJ, Lin Y-L, Leung ML, El-Naggar A, Creighton CJ, Suraokar MB, Wistuba I, Flores ER (2010) TAp63 suppresses metastasis through coordinate regulation of Dicer and miRNAs. *Nature* 467: 986-990.
- Sulik, K.K. dan Dehart, D.B. (1988) Retinoic-acid-induced limb malformations resulting from apical ectodermal ridge cell death. *Teratology*, 37, 527–537.
- Ugur SA dan Tolun A (2008) Homozygous WNT10b mutation and complex inheritance in split-hand/foot malformation. *Hum. Molec. Genet.* 17: 2644-2653.
- van Bookhoven H, Hamel BCJ, Bamshad M, Sangiorgi E, Gurrieri F, Duijf PHG, Vanmolkot KRJ, van Beusekom E, van Beersum SEC, Celli J, Merkx GFM, Tenconi R, Fryns JP, Verloes A, Newbury-Ecob RA, Raas-Rotschild A, Majewski F, Beemer FA, Janecke A, Chitayat D, Crisponi G, Kayserili H, Yates JRW, Neri G, Brunner HG (2001) *p63* Gene Mutations in EEC Syndrome, Limb-Mammary Syndrome, and Isolated Split Hand-Split Foot Malformation Suggest a Genotype-Phenotype Correlation. *Am J Hum Genet* 69:481-492.
- Yang A, Kaghad M, Wang Y, Gillett E, Fleming MD, Dotsch V, Andrews NC, Caput D, McKeon F (1998) p63, a p53 homolog at 3q27-29, encodes multiple products with transactivating, death-inducing, and dominant-negative activities. *Molec. Cell* 2: 305-316.