

## BIBIOGRAPHY

Aldred MJ, Crawford PJ (1995). Amelogenesis imperfecta--towards a new classification. *Oral Dis* 1(1):2-5.

Aldred MJ, Savarirayan R, Crawford PJ (2003). Amelogenesis imperfecta: a classification and catalogue for the 21st century. *Oral Dis* 9(1):19-23.

Aren G, Ozdemir D, Firatli S, Uygur C, Sepet E, Firatli E (2003). Evaluation of oral and systemic manifestations in an amelogenesis imperfecta population. *J Dent* 31(8):585-91.

Atasu M, Biren S, Mumcu G (1999). Hypocalcification type amelogenesis imperfecta in permanent dentition in association with heavily worn primary teeth, gingival hyperplasia, hypodontia and impacted teeth. *J Clin Pediatr Dent* 23(2):117-21.

Backman B, Holm AK (1986). Amelogenesis imperfecta: prevalence and incidence in a northern Swedish county. *Community Dent Oral Epidemiol* 14(1):43-7.

Backman B, Anneroth G (1989). Microradiographic study of amelogenesis imperfecta. *Scand J Dent Res* 97(4):316-29.

Backman B (1997). Inherited enamel defects. *Ciba Found Symp* 205(175-82; discussion 183-6).

Bartlett JD, Simmer JP, Xue J, Margolis HC, Moreno EC (1996). Molecular cloning and mRNA tissue distribution of a novel matrix metalloproteinase isolated from porcine enamel organ. *Gene* 183(1-2):123-8.

Bartlett JD, Ryu OH, Xue J, Simmer JP, Margolis HC (1998). Enamelysin mRNA displays a developmentally defined pattern of expression and encodes a protein which degrades amelogenin. *Connect Tissue Res* 39(1-3):101-9; discussion 141-9.

Bartlett JD, Simmer JP (1999). Proteinases in developing dental enamel. *Crit Rev Oral Biol Med* 10(4):425-41.

Begue-Kirn C, Krebsbach PH, Bartlett JD, Butler WT (1998). Dentin sialoprotein, dentin phosphoprotein, enamelysin and ameloblastin: tooth-specific molecules that are distinctively expressed during murine dental differentiation. *Eur J Oral Sci* 106(5):963-70.

Brennan MT, O'Connell BC, Rams TE, O'Connell AC (1999). Management of gingival overgrowth associated with generalized enamel defects in a child. *J Clin Pediatr Dent* 23(2):97-101.

Cerny R, Slaby I, Hammarstrom L, Wurtz T (1996). A novel gene expressed in rat ameloblasts codes for proteins with cell binding domains. *J Bone Miner Res* 11(7):883-91.

Chosack A, Eidelman E, Wisotski I, Cohen T (1979). Amelogenesis imperfecta among Israeli Jews and the description of a new type of local hypoplastic autosomal recessive amelogenesis imperfecta. *Oral Surg Oral Med Oral Pathol* 47(2):148-56.

Collier PM, Sauk JJ, Rosenbloom SJ, Yuan ZA, Gibson CW (1997). An amelogenin gene defect associated with human X-linked amelogenesis imperfecta. *Arch Oral Biol* 42(3):235-42.

Collins MA, Mauriello SM, Tyndall DA, Wright JT (1999). Dental anomalies associated with amelogenesis imperfecta: a radiographic assessment. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 88(3):358-64.

Crawford PJ, Aldred M, Bloch-Zupan A (2007). Amelogenesis imperfecta. *Orphanet J Rare Dis* 2(17).

Deutsch D, Palmon A, Fisher LW, Kolodny N, Termine JD, Young MF (1991). Sequencing of bovine enamelin ("tuftelin") a novel acidic enamel protein. *J Biol Chem* 266(24):16021-8.

Deutsch D, Catalano-Sherman J, Dafni L, David S, Palmon A (1995a). Enamel matrix proteins and ameloblast biology. *Connect Tissue Res* 32(1-4):97-107.

Deutsch D, Palmon A, Dafni L, Catalano-Sherman J, Young MF, Fisher LW (1995b). The enamelin (tuftelin) gene. *Int J Dev Biol* 39(1):135-43.

Feller L, Jadwat Y, Bouckaert M, Buskin A, Raubenheimer EJ (2006). Enamel dysplasia with odontogenic fibroma-like hamartomas: review of the literature and report of a case. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 101(5):620-4.

Fincham AG, Moradian-Oldak J, Simmer JP, Sarte P, Lau EC, Diekwisch T, et al. (1994). Self-assembly of a recombinant amelogenin protein generates supramolecular structures. *J Struct Biol* 112(2):103-9.

Fincham AG, Moradian-Oldak J, Diekwisch TG, Lyaruu DM, Wright JT, Bringas P, Jr., et al. (1995). Evidence for amelogenin "nanospheres" as functional components of secretory-stage enamel matrix. *J Struct Biol* 115(1):50-9.

Fincham AG, Simmer JP (1997). Amelogenin proteins of developing dental enamel. *Ciba Found Symp* 205(118-30; discussion 130-4).

Fong CD, Hammarstrom L (2000). Expression of amelin and amelogenin in epithelial root sheath remnants of fully formed rat molars. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 90(2):218-23.

Fukae M, Tanabe T, Murakami C, Dohi N, Uchida T, Shimizu M (1996). Primary structure of the porcine 89-kDa enamelin. *Adv Dent Res* 10(2):111-8.

Fukumoto S, Kiba T, Hall B, Iehara N, Nakamura T, Longenecker G, et al. (2004). Ameloblastin is a cell adhesion molecule required for maintaining the differentiation state of ameloblasts. *J Cell Biol* 167(5):973-83.

Gopinath VK, Al-Salihi KA, Yean CY, Ann MC, Ravichandran M (2004). Amelogenesis imperfecta: enamel ultra structure and molecular studies. *J Clin Pediatr Dent* 28(4):319-22.

Gopinath VK, Yoong TP, Yean CY, Ravichandran M (2008). Identifying polymorphism in enamelin gene in amelogenesis imperfecta (AI). *Arch Oral Biol* 53(10):937-40.

Greene SR, Yuan ZA, Wright JT, Amjad H, Abrams WR, Buchanan JA, et al. (2002). A new frameshift mutation encoding a truncated amelogenin leads to X-linked amelogenesis imperfecta. *Arch Oral Biol* 47(3):211-7.

Gutierrez SJ, Chaves M, Torres DM, Briceno I (2007). Identification of a novel mutation in the enamalin gene in a family with autosomal-dominant amelogenesis imperfecta. *Arch Oral Biol* 52(5):503-6.

Hart PS, Aldred MJ, Crawford PJ, Wright NJ, Hart TC, Wright JT (2002). Amelogenesis imperfecta phenotype-genotype correlations with two amelogenin gene mutations. *Arch Oral Biol* 47(4):261-5.

Hart PS, Michalec MD, Seow WK, Hart TC, Wright JT (2003a). Identification of the enamelin (g.8344delG) mutation in a new kindred and presentation of a standardized ENAM nomenclature. *Arch Oral Biol* 48(8):589-96.

Hart PS, Hart TC, Michalec MD, Ryu OH, Simmons D, Hong S, et al. (2004). Mutation in kallikrein 4 causes autosomal recessive hypomaturation amelogenesis imperfecta. *J Med Genet* 41(7):545-9.

Hart PS, Becerik S, Cogulu D, Emingil G, Ozdemir-Ozenen D, Han ST, et al. (2009). Novel FAM83H mutations in Turkish families with autosomal dominant hypocalcified amelogenesis imperfecta. *Clin Genet* 75(4):401-4.

Hart TC, Hart PS, Gorry MC, Michalec MD, Ryu OH, Uygur C, et al. (2003b). Novel ENAM mutation responsible for autosomal recessive amelogenesis imperfecta and localised enamel defects. *J Med Genet* 40(12):900-6.

Health Grades Inc. "Statistics by country for amelogenesis imperfecta." [Online]. Available [http://www.wrongdiagnosis.com/a/amelogenesis\\_imperfecta/stats-country.htm#extrapwarning](http://www.wrongdiagnosis.com/a/amelogenesis_imperfecta/stats-country.htm#extrapwarning) (26 January 2011).

Hirschberg A, Buchner A, Dayan D (1996). The central odontogenic fibroma and the hyperplastic dental follicle: study with Picrosirius red and polarizing microscopy. *J Oral Pathol Med* 25(3):125-7.

Hu CC, Ryu OH, Qian Q, Zhang CH, Simmer JP (1997). Cloning, characterization, and heterologous expression of exon-4-containing amelogenin mRNAs. *J Dent Res* 76(2):641-7.

Hu CC, Hart TC, Dupont BR, Chen JJ, Sun X, Qian Q, et al. (2000a). Cloning human enamelin cDNA, chromosomal localization, and analysis of expression during tooth development. *J Dent Res* 79(4):912-9.

Hu JC, Zhang C, Sun X, Yang Y, Cao X, Ryu O, et al. (2000b). Characterization of the mouse and human PRSS17 genes, their relationship to other serine proteases, and the expression of PRSS17 in developing mouse incisors. *Gene* 251(1):1-8.

Hu JC, Sun X, Zhang C, Simmer JP (2001a). A comparison of enamelin and amelogenin expression in developing mouse molars. *Eur J Oral Sci* 109(2):125-32.

Hu JC, Zhang CH, Yang Y, Karrman-Mardh C, Forsman-Semb K, Simmer JP (2001b). Cloning and characterization of the mouse and human enamelin genes. *J Dent Res* 80(3):898-902.

Hu JC, Sun X, Zhang C, Liu S, Bartlett JD, Simmer JP (2002). Enamelysin and kallikrein-4 mRNA expression in developing mouse molars. *Eur J Oral Sci* 110(4):307-15.

Hu JC, Yamakoshi Y (2003). Enamelin and autosomal-dominant amelogenesis imperfecta. *Crit Rev Oral Biol Med* 14(6):387-98.

Hu JC, Yamakoshi Y, Yamakoshi F, Krebsbach PH, Simmer JP (2005). Proteomics and genetics of dental enamel. *Cells Tissues Organs* 181(3-4):219-31.

Hu JC, Chun YH, Al Hazzazi T, Simmer JP (2007). Enamel formation and amelogenesis imperfecta. *Cells Tissues Organs* 186(1):78-85.

Hyun HK, Lee SK, Lee KE, Kang HY, Kim EJ, Choung PH, et al. (2009). Identification of a novel FAM83H mutation and microhardness of an affected molar in autosomal dominant hypocalcified amelogenesis imperfecta. *Int Endod J* 42(11):1039-43.

Inai T, Kukita T, Ohsaki Y, Nagata K, Kukita A, Kurisu K (1991). Immunohistochemical demonstration of amelogenin penetration toward the dental pulp in the early stages of ameloblast development in rat molar tooth germs. *Anat Rec* 229(2):259-70.

Kaffe I, Buchner A (1994). Radiologic features of central odontogenic fibroma. *Oral Surg Oral Med Oral Pathol* 78(6):811-8.

Kang HY, Seymen F, Lee SK, Yildirim M, Tuna EB, Patir A, et al. (2009). Candidate gene strategy reveals ENAM mutations. *J Dent Res* 88(3):266-9.

Kida M, Ariga T, Shirakawa T, Oguchi H, Sakiyama Y (2002). Autosomal-dominant hypoplastic form of amelogenesis imperfecta caused by an enamelin gene mutation at the exon-intron boundary. *J Dent Res* 81(11):738-42.

Kida M, Sakiyama Y, Matsuda A, Takabayashi S, Ochi H, Sekiguchi H, et al. (2007). A novel missense mutation (p.P52R) in amelogenin gene causing X-linked amelogenesis imperfecta. *J Dent Res* 86(1):69-72.

Kim JW, Simmer JP, Hu YY, Lin BP, Boyd C, Wright JT, et al. (2004). Amelogenin p.M1T and p.W4S mutations underlying hypoplastic X-linked amelogenesis imperfecta. *J Dent Res* 83(5):378-83.

Kim JW, Seymen F, Lin BP, Kiziltan B, Gencay K, Simmer JP, et al. (2005a). ENAM mutations in autosomal-dominant amelogenesis imperfecta. *J Dent Res* 84(3):278-82.

Kim JW, Simmer JP, Hart TC, Hart PS, Ramaswami MD, Bartlett JD, et al. (2005b). MMP-20 mutation in autosomal recessive pigmented hypomaturational amelogenesis imperfecta. *J Med Genet* 42(3):271-5.

Kim JW, Simmer JP, Lin BP, Seymen F, Bartlett JD, Hu JC (2006). Mutational analysis of candidate genes in 24 amelogenesis imperfecta families. *Eur J Oral Sci* 114 Suppl 1(3-12; discussion 39-41, 379.

Kim JW, Lee SK, Lee ZH, Park JC, Lee KE, Lee MH, et al. (2008). FAM83H mutations in families with autosomal-dominant hypocalcified amelogenesis imperfecta. *Am J Hum Genet* 82(2):489-94.

Kindelan SA, Brook AH, Gangemi L, Lench N, Wong FS, Fearn J, et al. (2000). Detection of a novel mutation in X-linked amelogenesis imperfecta. *J Dent Res* 79(12):1978-82.

Lagerstrom-Fermer M, Landegren U (1995). Understanding enamel formation from mutations causing X-linked amelogenesis imperfecta. *Connect Tissue Res* 32(1-4):241-6.

Lee MJ, Lee SK, Lee KE, Kang HY, Jung HS, Kim JW (2009). Expression patterns of the Fam83h gene during murine tooth development. *Arch Oral Biol* 54(9):846-50.

Lee SK, Hu JC, Bartlett JD, Lee KE, Lin BP, Simmer JP, et al. (2008). Mutational spectrum of FAM83H: the C-terminal portion is required for tooth enamel calcification. *Hum Mutat* 29(8):E95-E99.

Lench NJ, Brook AH, Winter GB (1994). SSCP detection of a nonsense mutation in exon 5 of the amelogenin gene (AMGX) causing X-linked amelogenesis imperfecta (AIH1). *Hum Mol Genet* 3(5):827-8.

Lench NJ, Winter GB (1995). Characterisation of molecular defects in X-linked amelogenesis imperfecta (AIH1). *Hum Mutat* 5(3):251-9.



Llano E, Pendas AM, Knauper V, Sorsa T, Salo T, Salido E, et al. (1997). Identification and structural and functional characterization of human enamelysin (MMP-20). *Biochemistry* 36(49):15101-8.

Macedo GO, Tunes RS, Motta AC, Passador-Santos F, Grisi MM, Souza SL, et al. (2005). Amelogenesis imperfecta and unusual gingival hyperplasia. *J Periodontol* 76(9):1563-6.

Mardh CK, Backman B, Holmgren G, Hu JC, Simmer JP, Forsman-Semb K (2002). A nonsense mutation in the enamelin gene causes local hypoplastic autosomal dominant amelogenesis imperfecta (AIH2). *Hum Mol Genet* 11(9):1069-74.

Martelli-Junior H, Bonan PR, Dos Santos LA, Santos SM, Cavalcanti MG, Coletta RD (2008). Case reports of a new syndrome associating gingival fibromatosis and dental abnormalities in a consanguineous family. *J Periodontol* 79(7):1287-96.

Masuya H, Shimizu K, Sezutsu H, Sakuraba Y, Nagano J, Shimizu A, et al. (2005). Enamelin (Enam) is essential for amelogenesis: ENU-induced mouse mutants as models for different clinical subtypes of human amelogenesis imperfecta (AI). *Hum Mol Genet* 14(5):575-83.

Nagano T, Oida S, Ando H, Gomi K, Arai T, Fukae M (2003). Relative levels of mRNA encoding enamel proteins in enamel organ epithelia and odontoblasts. *J Dent Res* 82(12):982-6.

Nakamura M, Bringas P, Jr., Nanci A, Zeichner-David M, Ashdown B, Slavkin HC (1994). Translocation of enamel proteins from inner enamel epithelia to odontoblasts during mouse tooth development. *Anat Rec* 238(3):383-96.

Nanci A (2008). Ten Cate's Oral Histology: Development, Structure, and Function. 7th ed.: Mosby

Ooya K, Nalbandian J, Noikura T (1988). Autosomal recessive rough hypoplastic amelogenesis imperfecta. A case report with clinical, light microscopic, radiographic, and electron microscopic observations. *Oral Surg Oral Med Oral Pathol* 65(4):449-58.

Ozdemir D, Hart PS, Firatli E, Aren G, Ryu OH, Hart TC (2005a). Phenotype of ENAM mutations is dosage-dependent. *J Dent Res* 84(11):1036-41.

Ozdemir D, Hart PS, Ryu OH, Choi SJ, Ozdemir-Karatas M, Firatli E, et al. (2005b). MMP20 active-site mutation in hypomaturational amelogenesis imperfecta. *J Dent Res* 84(11):1031-5.

Papagerakis P, MacDougall M, Hotton D, Bailleul-Forestier I, Oboeuf M, Berdal A (2003). Expression of amelogenin in odontoblasts. *Bone* 32(3):228-40.

Pavlic A, Petelin M, Battelino T (2007). Phenotype and enamel ultrastructure characteristics in patients with ENAM gene mutations g.13185-13186insAG and 8344delG. *Arch Oral Biol* 52(3):209-17.

Peters E, Cohen M, Altini M (1992). Rough hypoplastic amelogenesis imperfecta with follicular hyperplasia. *Oral Surg Oral Med Oral Pathol* 74(1):87-92.

Poulsen S, Gjørup H, Haubek D, Haukali G, Hintze H, Lovschall H, et al. (2008). Amelogenesis imperfecta - a systematic literature review of associated dental and oro-facial abnormalities and their impact on patients. *Acta Odontol Scand* 66(4):193-9.

Rajpar MH, Harley K, Laing C, Davies RM, Dixon MJ (2001). Mutation of the gene encoding the enamel-specific protein, enamelin, causes autosomal-dominant amelogenesis imperfecta. *Hum Mol Genet* 10(16):1673-7.

Raubenheimer EJ, Noffke CE (2002). Central odontogenic fibroma-like tumors, hypodontia, and enamel dysplasia: review of the literature and report of a case. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 94(1):74-7.

Ravassipour DB, Hart PS, Hart TC, Ritter AV, Yamauchi M, Gibson C, et al. (2000). Unique enamel phenotype associated with amelogenin gene (AMELX) codon 41 point mutation. *J Dent Res* 79(7):1476-81.

Rawlings ND, Tolle DP, Barrett AJ (2004). MEROPS: the peptidase database. *Nucleic Acids Res* 32(Database issue):D160-4.

Robinson C, Shore RC, Kirkham J, Stonehouse NJ (1990). Extracellular processing of enamel matrix proteins and the control of crystal growth. *J Biol Buccale* 18(4):355-61.

Roquebert D, Champsaur A, Gil del Real P, Prasad H, Rohrer MD, Pintado M, et al. (2008). Amelogenesis imperfecta, rough hypoplastic type, dental follicular hamartomas and gingival hyperplasia: report of a case from Central America and review of the literature. *Oral Surg Oral Med Oral Pathol Oral Radiol Endod* 106(1):92-8.

Ryu O, Hu JC, Yamakoshi Y, Villemain JL, Cao X, Zhang C, et al. (2002). Porcine kallikrein-4 activation, glycosylation, activity, and expression in prokaryotic and eukaryotic hosts. *Eur J Oral Sci* 110(5):358-65.

Santos MC, Hart PS, Ramaswami M, Kanno CM, Hart TC, Line SR (2007). Exclusion of known gene for enamel development in two Brazilian families with amelogenesis imperfecta. *Head Face Med* 3(8).

Seow WK (1993). Clinical diagnosis and management strategies of amelogenesis imperfect variants. *Pediatr Dent* 15(6):384-93.

Simmer JP, Hu JC (2001). Dental enamel formation and its impact on clinical dentistry. *J Dent Educ* 65(9):896-905.

Slavkin HC, Bessem C, Bringas P, Jr., Zeichner-David M, Nanci A, Snead ML (1988). Sequential expression and differential function of multiple enamel proteins during fetal, neonatal, and early postnatal stages of mouse molar organogenesis. *Differentiation* 37(1):26-39.

Smith CE, Warshawsky H (1977). Quantitative analysis of cell turnover in the enamel organ of the rat incisor. Evidence for ameloblast death immediately after enamel matrix secretion. *Anat Rec* 187(1):63-98.

Smith CE, McKee MD, Nanci A (1987). Cyclic induction and rapid movement of sequential waves of new smooth-ended ameloblast modulation bands in rat incisors as visualized by polychrome fluorescent labeling and GBHA-staining of maturing enamel. *Adv Dent Res* 1(2):162-75.

Smith CE, Dahan S, Fazel A, Lai W, Nanci A (1992). Correlated biochemical and radioautographic studies of protein turnover in developing rat incisor enamel following pulse-chase labeling with L-[<sup>35</sup>S]- and L-[methyl-<sup>3</sup>H]-methionine. *Anat Rec* 232(1):1-14.

Smith CE, Nanci A (1995). Overview of morphological changes in enamel organ cells associated with major events in amelogenesis. *Int J Dev Biol* 39(1):153-61.

Snead ML, Lau EC, Zeichner-David M, Fincham AG, Woo SL, Slavkin HC (1985). DNA sequence for cloned cDNA for murine amelogenin reveal the amino acid sequence for enamel-specific protein. *Biochem Biophys Res Commun* 129(3):812-8.

Snead ML, Luo W, Lau EC, Slavkin HC (1988). Spatial- and temporal-restricted pattern for amelogenin gene expression during mouse molar tooth organogenesis. *Development* 104(1):77-85.

Stephanopoulos G, Garefalaki ME, Lyroudia K (2005). Genes and related proteins involved in amelogenesis imperfecta. *J Dent Res* 84(12):1117-26.

Sukigushi H KM, Yakushiji M (2001). DNA diagnosis of X-linked amelogenesis imperfecta using PCR detection method of the human amelogenin gene. *Dent Jpn* 37(109-112).

van Heerden WF, Raubenheimer EJ, Dreyer AF, Benn AM (1990). Amelogenesis imperfecta: multiple impactions associated with odontogenic fibromas (WHO) type. *J Dent Assoc S Afr* 45(11):467-71.

Warshawsky H (1978). A freeze-fracture study of the topographic relationship between inner enamel-secretory ameloblasts in the rat incisor. *Am J Anat* 152(2):153-207.

Witkop CJ (1957). Hereditary defects in enamel and dentin. *Acta Genet Stat Med* 7(1):236-9.

Witkop CJ, Jr. (1967). Partial expression of sex-linked recessive amelogenesis imperfecta in females compatible with the Lyon hypothesis. *Oral Surg Oral Med Oral Pathol* 23(2):174-82.

Witkop CJ, Jr. (1988). Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: problems in classification. *J Oral Pathol* 17(9-10):547-53.

Wright JT, Hart PS, Aldred MJ, Seow K, Crawford PJ, Hong SP, et al. (2003). Relationship of phenotype and genotype in X-linked amelogenesis imperfecta. *Connect Tissue Res* 44 Suppl 1(72-8).

Wright JT (2006). The molecular etiologies and associated phenotypes of amelogenesis imperfecta. *Am J Med Genet A* 140(23):2547-55.

Zeichner-David M, Diekwisch T, Fincham A, Lau E, MacDougall M, Moradian-Oldak J, et al. (1995). Control of ameloblast differentiation. *Int J Dev Biol* 39(1):69-92.

Zeichner-David M, Vo H, Tan H, Diekwisch T, Berman B, Thiemann F, et al. (1997). Timing of the expression of enamel gene products during mouse tooth development. *Int J Dev Biol* 41(1):27-38.