

Table SI. COL7A1 glycine substitution mutations with dominant inheritance

Diagnosis	Exon	Glycine Substitution	cDNA	Reference
DDEB-BDN	42	p.Gly1483Asp	c.4448G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB	45	p.Gly1522Glu	c.4565G>A	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
DDEB-gen	48	p.Gly1557Arg	c.4669G>A	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
DDEB-na	50	p.Gly1595Arg	c.4783G>C	Sato-Matsumura et al. Arch Dermatol 2002; 138: 269–271.
DDEB-gen	53	p.Gly1649Arg	c.4945G>C	This paper
DDEB-pr, DDEB-gen	56	p.Gly1700Asp	c.5099G>A	Deng et al. J Dermatol Sci 2008; 49: 166–169.
DDEB-gen	58	p.Gly1734Asp	c.5201G>A	This paper
DDEB-pr	59	p.Gly1755Asp ^a	c.5264G>C	Posteraro et al. Biochem Biophys Res Commun 2005; 338: 1391–1401.
DDEB-gen	59	p.Gly1755Val	c.5264G>T	This paper
DDEB-gen	60	p.Gly1764Asp	c.5291G>A	This paper
DDEB-na, DDEB-pt	61	p.Gly1770Ser	c.5308G>A	This paper
DDEB-pr	61	p.Gly1770Asp	c.5309G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	61	p.Gly1773Asp	c.5318G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB	61	p.Gly1776Arg	c.5326G>A	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
DDEB-gen	61	p.Gly1776Trp ^a	c.5326G>T	Bursztejn et al. Ann Dermatol Venereol 2008; 135: 195–199.
DDEB-gen	61	p.Gly1776Glu	c.5327G>A	This paper
DDEB-pr	61	p.Gly1791Glu	c.5372G>A	Mellerio et al. J Invest Dermatol 1999; 112: 984–987.
DDEB-na	63	p.Gly1815Arg	c.5443G>A	Sato-Matsumura et al. Arch Dermatol 2002; 138: 269–271.
DDEB-pr	66	p.Gly1860Arg	c.5578G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-pr	69	p.Gly1913Arg	c.5737G>C	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	73	p.Gly2003Arg	c.6007G>A	Christiano et al. J Invest Dermatol 1996; 106: 1340–1342.
DDEB-gen	73	p.Gly2003Glu	c.6008G>A	This paper
DDEB-gen	73	p.Gly2006Ser ^a	c.6016G>A	Mallipeddi et al. Br J Dermatol 2003; 149: 810–818.
DDEB-gen	73	p.Gly2006Asp	c.6017G>A	Hammami-Hauasli et al. J Biol Chem 1998; 273: 1928–1934.
DDEB	73	p.Gly2006Ala	c.6018G>C	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
DDEB-gen	73	p.Gly2009Ala	c.6026G>C	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB	73	p.Gly2012Ser ^a	c.6034G>A	Gardella et al. J Invest Dermatol 2002; 119: 1456–1462.
DDEB-gen	73	p.Gly2012Asp ^a	c.6035G>A	Matsuba et al. Clin Exp Dermatol 2002; 27: 56–58.
DDEB-gen	73	p.Gly2015Glu	c.6044G>A	Hammami-Hauasli et al. J Biol Chem 1998; 273: 1928–1934.
DDEB-gen	73	p.Gly2028Arg	c.6082G>A	Lee et al. Arch Dermatol Res 2000; 292: 159–163.
DDEB-gen	73	p.Gly2028Trp	c.6082G>T	Varki et al. J Med Genet 2007; 44: 181–192.
DDEB-gen	73	p.Gly2028Ala	c.6083G>C	Murata et al. Arch Dermatol Res 2000; 292: 477–481.
DDEB-gen	73	p.Gly2028Glu	c.6083G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	73	p.Gly2034Arg	c.6100G>A	Kon et al. J Invest Dermatol 1997; 109: 684–687.
DDEB-gen	73	p.Gly2034Trp	c.6100G>T	Rouan et al. J Invest Dermatol 1998; 111: 1210–1213.
DDEB	73	p.Gly2034Glu	c.6101G>A	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
DDEB-gen	73	p.Gly2037Arg	c.6109G>A	Iwata et al. J Dermatol 2006; 33: 550–556.
DDEB-gen	73	p.Gly2037Glu	c.6110G>A	Jonkman et al. J Invest Dermatol 1999; 112: 815–817.
DDEB-gen	73	p.Gly2040Ser	c.6118G>A	Christiano et al. Proc Natl Acad Sci U S A 1994; 91: 3549–3553.
DDEB	73	p.Gly2040Asp	c.6119G>A	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
DDEB-gen	73	p.Gly2040Val ^a	c.6119G>T	Rouan et al. J Invest Dermatol 1998; 111: 1210–1213.
DDEB	73	p.Gly2043Arg ^a	c.6127G>A	Christiano et al. J Invest Dermatol 1995; 104: 438–440.
DDEB-gen	73	p.Gly2043Trp ^a	c.6127G>T	Mecklenbeck et al. J Invest Dermatol 1999; 112: 398–400.
DDEB	73	p.Gly2046Val	c.6137G>T	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
DDEB-gen	73	p.Gly2046Asp	c.6137G>A	This paper
DDEB-gen	73	p.Gly2055Glu	c.6164G>A	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
DDEB-pt	73	p.Gly2059Glu ^b	c.6137G>A ^c	Hamada et al. J Dermatol Sci 2009; 54: 212–214.
DDEB-gen	74	p.Gly2061Val	c.6182G>T	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
DDEB-gen	74	p.Gly2064Arg	c.6190G>A	Rouan et al. J Invest Dermatol 1998; 111: 1210–1213.
DDEB-gen	74	p.Gly2064Val	c.6191G>T	Kern et al. Br J Dermatol 2009; 161: 1089–1097.
DDEB	74	p.Gly2064Glu	c.6191G>A	Sawamura et al. J Hum Genet 2005; 50: 543–546.
DDEB	74	p.Gly2067Arg ^a	c.6199G>A	Posteraro et al. Biochem Biophys Res Commun 2005; 338: 1391–1401.
DDEB-gen	74	p.Gly2067Ala	c.6200G>C	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	74	p.Gly2070Arg	c.6208G>A	Zhang et al. Clin Exp Dermatol 2003; 28: 437–439.
DDEB	74	p.Gly2070Glu	c.6209G>A	Gardella et al. J Invest Dermatol 2002; 119: 1456–1462.
DDEB-pr	75	p.Gly2073Val	c.6218G>T	Drera et al. Clin Genet 2006; 70: 339–347
DDEB-gen	75	p.Gly2076Asp ^a	c.6227G>A	Kon et al. J Invest Dermatol 1997; 109: 684–687.
DDEB	75	p.Gly2079Arg	c.6235G>A	Christiano et al. Exp Dermatol 1999; 8: 146–152.
DDEB-gen	75	p.Gly2079Glu ^a	c.6236G>A	Kon et al. J Invest Dermatol 1997; 108: 224–228.
DDEB-gen	75	p.Gly2079Val	c.6236G>T	Kern et al. Br J Dermatol 2009; 161: 1089–1097.
DDEB-pr	79	p.Gly2159Glu	c.6476G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.

DDEB-BDN	81	p.Gly2186Glu	c.6557G>A	This paper
DDEB-gen	83	p.Gly2207Arg	c.6619G>A	Kon et al. J Invest Dermatol 1998; 111: 534–537.
DDEB-pr	83	p.Gly2210Val	c.6629G>T	Dang et al. J Dermatol Sci 2007; 46: 169–178.
DDEB-pr	83	p.Gly2213Arg	c.6637G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	84	p.Gly2227Arg	c.6679G>A	This paper
DDEB-gen	84	p.Gly2230Glu	c.6689G>A	Kern et al. Br J Dermatol 2009; 161: 1089–1097.
DDEB-pr	84	p.Gly2233Glu	c.6698G>A	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-pr	85	p.Gly2239Asp	c.6716G>A	Tamai et al. J Invest Dermatol 1998; 110: 509.
DDEB-pr	85	p.Gly2239Val	c.6716G>T	This paper
DDEB-pr	85	p.Gly2242Arg	c.6724G>A	Lee et al. J Invest Dermatol 1997; 108: 947–949.
DDEB-pr	85	p.Gly2242Trp	c.6724G>T	Shi et al. Clin Exp Dermatol 2009; 34: e975–978.
DDEB-pr	85	p.Gly2242Glu	c.6725G>A	Tamai et al. J Invest Dermatol 1998; 110: 509.
DDEB-na	86	p.Gly2251Glu	c.6752G>A	Hammami-Hauasli et al. J Invest Dermatol 1998; 111: 1214–1219.
DDEB-gen	86	p.Gly2260Asp	c.6779G>A	This paper
DDEB-na	87	p.Gly2287Arg	c.6859G>A	Shimizu et al. J Invest Dermatol 1999; 113: 419–421.
DDEB	87	p.Gly2287Val	c.6860G>T	Posteraro et al. Biochem Biophys Res Commun 2005; 338: 1391–1401.
DDEB-pr	87	p.Gly2290Ala	c.6869G>C	Almaani et al. Acta Derm Venereol 2009; 89: 6–11.
DDEB-gen	91	p.Gly2347Arg ^a	c.7042G>C	Cserhalmi-Friedman et al. Exp Dermatol 1999; 8: 143–145.
DDEB-gen	91	p.Gly2351Arg	c.7051G>A	Christiano et al. Am J Hum Genet 1996; 58: 682–693.
DDEB-pr	92	p.Gly2360Arg	c.7078G>A	This paper
DDEB-pr	92	p.Gly2366Val	c.7097G>T	Chuang et al. Clin Exp Dermatol 2004; 29: 304–307.
DDEB-pr	93	p.Gly2369Ser	c.7105G>A	Mellerio et al. J Invest Dermatol 1999; 112: 984–987.
DDEB-gen	95	p.Gly2428Asp^a	c.7283G>A	This paper
DDEB-pr	100	p.Gly2508Asp	c.7523G>A	This paper
DDEB-pr	100	p.Gly2517Asp	c.7550G>A	This paper
DDEB-pt	105	p.Gly2623Cys	c.7867G>T	Christiano et al. Hum Mol Genet 1995; 4: 1579–1583.
DDEB-pr	105	p.Gly2623Val ^b	c.7868G>T	Schumann et al. Br J Dermatol 2008; 159: 464–469.
DDEB-pr	106	p.Gly2626Asp	c.7877G>A	Wang et al. J Dermatol Sci 2007; 46: 211–213.
DDEB	106	p.Gly2632Arg	c.7894G>A	Posteraro et al. Biochem Biophys Res Commun 2005; 338: 1391–1401.
DDEB-pr	108	p.Gly2680Asp	c.8039G>A	This paper
DDEB-pr	110	p.Gly2713Arg	c.8137G>C	Mellerio et al. J Invest Dermatol 1999; 112: 984–987.
DDEB-gen	110	p.Gly2713Asp	c.8138G>A	Rouan et al. J Invest Dermatol 1998; 111: 1210–1213.
DDEB-pr	110	p.Gly2719Asp	c.8156G>A	Riedi et al. Pediatr Dermatol 2009; 26: 115–117.
DDEB-gen	110	p.Gly2722Arg	c.8164G>C	This paper
DDEB-gen	110	p.Gly2743Arg	c.8227G>C	This paper

DDEB: dominant dystrophic epidermolysis bullosa; DDEB-BDN: bullous dermolysis of the newborn; DDEB-gen: generalized; DDEB-na: nails only; DDEB-pr: pruriginosa; DDEB-pt: pretibial; MS: missense; **bold**: new mutations.

^a*De novo* mutations. ^bp.Gly2059Glu, ^cc.6137G>A, but corrected nomenclature should be: p.Gly2058Glu, c.6173G>A, respectively.