Table SII. *COL7A1 glycine substitution mutations with recessive inheritance*

	Allele 1			Allele 2				
Diagnosis	Exon	Glycine Substitution	cDNA	Exon/Intron	cDNA	Consequence	Protein	Reference
RDEB-O	4	p.Gly150Arg	c.448G>A	IVS5	c.682+1G>A	SS	Predicted retention of intron 5 + p.Pro228fsX32	Drera et al. J Dermatol Sci 2009; 53: 222–225.
RDEB-sev gen	4	**p.Gly174Arg**	c.520G>A	4	c.520G>A	GS[a]	p.Gly174Arg	**This paper**
RDEB-O	32	p.Gly1314Arg	c.3940G>A	78	c.6422delG	PTC	p.Gly2141fsX64	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB	33	p.Gly1332Asp	c.3995G>A	ND	ND	ND	ND	Garcia et al. Hum Genet 2009; 126: 334–335.
RDEB-O	34	**p.Gly1338Arg**	c.4012G>A	IVS34	c.4048-1G>A	SS	Predicted skipping of exon 35 (in-frame; 24aa)	Van den Akker et al. J Dermatol Sci 2009; 56: 9–18.
RDEB-sev gen	34	p.Gly1338Val	c.4013G>T	ND	ND	ND	ND	Dang et al. J Dermatol Sci 2007; 46: 169–178.
RDEB-O	34	p.Gly1347Arg	c.4039G>C	70	c.5820G>A	SS	Predicted skipping of exon 70 (in-frame; 16aa)	Terracina et al. J Invest Dermatol 1998; 111: 744–750.
RDEB-O	34	p.Gly1347Trp	c.4039G>T	IVS30	c.3832-2A>G	SS	in-frame deletion	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-O	36	p.Gly1383Arg	c.4147G>A	80	c.6527insC	PTC	p.Pro2176fsX113	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-O	42	**p.Gly1483Asp**	c.4448G>A	42	c.4448G>A	GS	p.Gly1483Asp	**This paper**
RDEB-O	42	p.Gly1492Arg	c.4474G>A	4	c.497insA	PTC	p.Gln166fsX51	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-BDN	44	p.Gly1519Asp	c.4556G>A	86	c.6752G>A	GS	p.Gly2251Glu	Hammami-Hauasli et al. J Invest Dermatol 1998; 111: 1214–1219.
RDEB-O	45	p.Gly1522Arg	c.4564G>C	ND	ND	ND	ND	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-pr	48	**p.Gly1572Ala**	c.4715G>C	104	c.7786delG	PTC	p.Gly2596fsX49	**This paper**
RDEB-O	50	p.Gly1595Arg	c.4783G>C	115	c.8479C>T	PTC	p.Gln2827Stop	Sato-Matsumura et al. Arch Dermatol 2002; 138: 269–271.
RDEB-O	50	p.Gly1604Arg	c.4810G>A	72	c.5964insC	PTC	p.Gly1988fsX110	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-O	51	p.Gly1616Arg	c.4846G>A	51	c.4846G>A	GS	p.Gly1616Arg	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-O	53	p.Gly1652Arg	c.4954G>A	3	c.425A>G	SS	p.Lys142Arg	Cserhalmi-Friedman et al. Arch Dermatol Res 1997; 289: 640–645.
RDEB-O	53	p.Gly1655Gly	c.4965C>T[b]	74	c.6187C>T	MS	p.Arg2063Trp	Gardella et al. J Invest Dermatol 2002; 119: 1456–1462.
RDEB-O	54	p.Gly1664Ala	c.4991G>C	54	c.4991G>C or	GS	p.Gly1664Ala	Gardella et al. J Invest Dermatol 2002; 119: 1456–1462.
				4	c.497insA or	PTC	p.Gln166fsX51	
				3	c.425A>G	SS[c]	p.Lys142Arg	
RDEB-O	54	p.Gly1673Arg	c.5017G>A	ND	ND	ND	ND	Dang et al. J Dermatol Sci 2007; 46: 169–178.
RDEB-O	56	p.Gly1696Glu[d]	c.5090G>A	84	c.6691insC	PTC	p.Pro2231fsX58	Salas-Alanis et al. Int J Dermatol 2000; 39: 436–442.
RDEB-O	56	p.Gly1703Glu	c.5108G>A	69	c.5772delG	PTC	p.Glu1924fsX50	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-O	58	p.Gly1719Arg	c.5155G>C	58	c.5155G>C	GS	p.Gly1719Arg	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-sev gen	61	**p.Gly1770Ser**	c.5308G>A	61	c.5308G>A	GS[e]	p.Gly1770Ser	**This paper**
RDEB-O	61	p.Gly1782Arg	c.5344G>A	73	c.6081delC	PTC	p.Pro2027fsX178	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
RDEB	61	p.Gly1782Val	c.5345G>T	3	c.425A>G	SS[c]	p.Lys142Arg	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-O	63	p.Gly1812Arg	c.5434G>C	31	c.3857delA	PTC	p.Gln1286fsX38	Masunaga et al. J Invest Dermatol 2000; 114: 204–205.
RDEB-O	63	p.Gly1815Arg	c.5443G>A	70	c.5818delC	PTC	p.Pro1940fsX64	Sato-Matsumura et al. Arch Dermatol 2002; 138: 269–271.
RDEB-O	65	p.Gly1845Arg	c.5533G>A	3	c.425A>G	SS[c]	p.Lys142Arg	Jerábková et al. J Dermatol Sci 2010; 59: 136–140.
RDEB-sev gen	66	p.Gly1857Glu	c.5570G>A	66	c.5570G>A	GS	p.Gly1857Glu	Kern et al. Br J Dermatol 2009; 161: 1089–1097.
RDEB-i	68	p.Gly1907Glu	c.5720G>A	105	c.7805G>A	GS	p.Gly2602Glu	Van den Akker et al. J Dermatol Sci 2009; 56: 9–18.
RDEB-i	68	**p.Gly1907Asp**	c.5720-21 GA>AT	54	c.5047C>T	PTC	p.Arg1683Stop	**This paper**
RDEB-O	68	p.Gly1907Asp[f]	c.5723G>A	13	c.1732C>T or	PTC	p.Arg578Stop	Varki et al. J Med Genet 2007; 44: 181–192.
				76	c.6311del2	PTC	p.Ser2104fsX11	
RDEB-BDN	68	p.Gly1910Ser	c.5728G>A	IVS5	c.682+1G>A	SS	predicted retention of intron 5 + p.Pro228fsX32	Hashikawa et al. J Dermatol Sci 2009; 56: 66–68.
RDEB-sev gen	72	p.Gly1982Trp	c.5944G>T	5	c.553C>T	PTC	p.Arg185Stop	Hovnanian et al. Am J Hum Genet 1997; 61: 599–610.
RDEB-O	72	**p.Gly1982Glu**	c.5945G>A	12	c.1573C>T	PTC	p.Arg525Stop	**This paper**
RDEB-O	73	p.Gly2009Arg	c.6025G>A	115	c.8523del14	SS	predicted skipping of exon 115 (in-frame; 29aa)	Winberg et al. Hum Mol Genet 1997; 6: 1125–1135.
RDEB	73	p.Gly2025Ser	c.6073G>A	ND	ND	ND	ND	Pfendner et al. Prenat Diagn 2003; 23: 447–456.
RDEB-O	73	p.Gly2025Ala	c.6074G>C	105	c.7828C>A	PTC	p.Arg2610Stop	Hovnanian et al. Am J Hum Genet 1997; 61: 599–610.
RDEB-O	73	p.Gly2028Trp	c.6082G>T	117	c.8698del11	PTC	p.Ser2900fsX19	Varki et al. J Med Genet 2007; 44: 181–192.

RDEB- sev gen	73	p.Gly2028Arg	c.6082G>A	13	c.1661del57	PTC	NR	Varki et al. J Med Genet 2007; 44: 181–192.
RDEB- sev gen	73	p.Gly2031Ser	c.6080G>A[g]	73	c.6080G>A	GS	p.Gly2031Ser	Nordal et al. Br J Dermatol 2001; 144: 151–157.
RDEB-sev gen	73	p.Gly2049Glu	c.6146G>A	34	c.4027C>T	PTC	p.Arg1343Stop	Hovnanian et al. Am J Hum Genet 1997; 61: 599–610.
RDEB-O	73	p.Gly2058Ala	c.6173G>C	102	c.7621C>T	PTC	p.Arg2541Stop	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB-sev gen	74	p.Gly2061Glu	c.6182G>A	IVS116	c.8620+26G>A	SS	ND	Chao et al. J Formos Med Assoc 2007; 106: 86–91.
RDEB-O	75	p.Gly2073Asp[h]	c.6218G>A	13	c.1732C>T	PTC	p.Arg578Stop	Dunnill et al. J Invest Dermatol 1996; 107: 171–177.
RDEB-i	75	p.Gly2088Arg	c.6262 G>A	5	c.676 C>T	PTC	p.Arg226Stop	Chiaverini et al. J Invest Dermatol 2010; 130: 2508–2511.
RDEB-O	75	p.Gly2088Glu	c.6263G>A	116	c.8569G>T	PTC	p.Glu2857Stop	Suzuki et al. Br J Dermatol 2006; 155: 838–840.
RDEB-O	78	p.Gly2132Ser	c.6394G>A	IVS5	c.682+1G>A	SS	predicted retention of intron 5 + p.Pro228fsX32	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB-O	78	p.Gly2132Asp	c.6395G>A	58	c.5188C>T	PTC	p.Arg1730Stop	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-O	82	p.Gly2192Ser	c.6574G>A	73	c.6081delC	PTC	p.Pro2027fsX178	Whittock et al. J Invest Dermatol 1999; 113: 673–86.
RDEB-sev gen	82	p.Gly2204Ser	c.6610G>A	55	c.5066C>G	PTC	p.Ser1689XStop	Kim et al. J Dermatol Sci 2003; 33: 180–183.
RDEB-sev gen	83	p.Gly2210Val	c.6629G>T	113	c.8371C>T	MS	p.Arg2791Trp	Varki et al. J Med Genet 2007; 44: 181–192.
RDEB-O	83	**p.Gly2213Arg**	c.6637G>A	52	c.4918delG	PTC	p.Gly1640fsX69	**This paper**
RDEB-O	84	**p.Gly2221Arg**	c.6661G>A	49	c.4748delG	PTC	p.Gly1583fsX126	**This paper**
RDEB-O	84	p.Gly2221Ala	c.6662G>C	IVS106	c.7930-1G>C	SS	predicted skipping of exon 106 (in-frame, 18aa)	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-O	84	p.Gly2233Cys	c.6697G>T	84	c.6695C>G	MS	p.Pro2232Arg	Ryoo et al. J Dermatol Sci 2001; 26: 125–132.
RDEB-O	84	p.Gly2233Ser	c.6697G>A	ND	ND	ND	ND	Salas-Alanis et al. Int J Dermatol 2000; 39: 436–442.
RDEB- sev gen	86	p.Gly2251Ala/ p.Pro1458Leu[i]	c.6752G>C	103	c.7723G>A	GS	p.Gly2575Arg	Varki et al. J Med Genet 2007; 44: 181–192.
RDEB-BDN	86	p.Gly2251Glu	c.6752G>A	44	c.4556G>A	GS	p.Gly1519Asp	Hammami-Hauasli et al. J Invest Dermatol 1998; 111: 1214–1219.
RDEB-O	86	p.Gly2263Val	c.6788G>T	13	c.1732C>T	PTC	p.Arg578Stop	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-O	86	**p.Gly2272Ala**	c.6815G>C	21	ND	ND	ND	**This paper**
RDEB-O	87	p.Gly2287Arg	c.6859G>A	89	c.6946G>A	GS	p.Gly2316Arg	Shimizu et al. J Invest Dermatol 1999; 113: 419–421.
RDEB-O	87	p.Gly2296Glu	c.6887G>A	12	c.1573C>T	PTC	p.Arg525Stop	Jerábková et al. J Dermatol Sci 2010; 59: 136–140.
RDEB-O	89	p.Gly2316Arg	c.6946G>A	87	c.6859G>A	GS	p.Gly2287Arg	Shimizu et al. J Invest Dermatol 1999; 113: 419–421.
RDEB-sev gen	91	p.Gly2351Arg	c.7051G>A	56	c.5103delCCinsG	PTC	p.Gly1701fsX8	Christiano et al. Am J Hum Genet 1996; 58: 682–693.
RDEB-O	92	p.Gly2357Ser	c.7069G>A	ND	ND	ND	ND	Varki et al. J Med Genet 2007; 44: 181–192.
RDEB-O	92	**p.Gly2357Arg**	c.7069G>C	IVS64	c.5487+4delAGTG	PTC	in-frame deletion	**This paper**
RDEB-O	92	p.Gly2366Cys	c.7096G>T	70	c.5818delC	PTC	p.Pro1940fsX64	Sawamura et al. J Hum Genet 2005; 50: 543–546.
RDEB-O	92	p.Gly2366Ser	c.7096G>A	74	c.6187C>T	MS	p.Arg2063Trp	Hashimoto et al. Exp Dermatol 1999; 8: 140–142.
RDEB-sev gen	92	p.Gly2366Asp	c.7097G>A	80	c.6527insC	PTC	p.Prol2176fsX113	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-sev gen	93	**p.Gly2369Ser**	c.7105G>A	93	c.7105G>A	GS	p.Gly2369Ser	**This paper**
RDEB-BDN	93	**p.Gly2372Val**	c.7115G>T	9	c.7115G>T	GS	p.Gly2372Val	**This paper**
RDEB-sev gen	93	p.Gly2375Ser[j]	NR	55	c.5097G>A	SS	in-frame deletion	Fassihi et al. J Dermatol Sci 2006; 42: 241–248.
RDEB-sev gen	94	p.Gly2395Asp	c.7184G>A	2	c.154delG	del	In-frame deletion	Mayama et al. J Invest Dermatol 1999; 112: 568.
RDEB-O	94	p.Gly2413Arg	c.7237G>A	20	c.2699G>A	PTC	p.Trp900Stop	Van den Akker et al. J Dermatol Sci 2009; 56: 9–18.
RDEB	94	p.Gly2413Glu	c.7238G>A	3	c.425A>G	SS[c]	p.Lys142Arg	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-O	94	p.Gly2422Val	c.7265G>T	94	c.7265G>T	GS	p.Gly2422Val	Kraemer et al. Eur J Dermatol 2006; 16: 615–619.
RDEB-O	95	p.Gly2434Arg	c.7300G>A	80	c.6527insC	PTC	p.Prol2176fsX113	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-sev gen	96	p.Gly2449Arg	c.7345G>A	96	c.7345G>A	GS	p.Gly2449Arg	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB-i	97	p.Gly2472Asp	c.7415G>A	14	c.1874del2	PTC	p.Ser625fsX4	Chiaverini et al. J Invest Dermatol 2010; 130: 2508–2511.
RDEB-pt	101	p.Gly2520Val	c.7559G>T	110	c.8209G>C	GS	p.Gly2737Arg	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-O	101	p.Gly2533Asp	c.7598G>A	26	c.3474delA	PTC	p.Pro1158fsX2	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB-sev gen	102	p.Gly2557Arg	c.7669G>A	34	c.4027C>T	PTC	p.Arg1343Stop	Jerábková et al. J Dermatol Sci 2010; 59: 136–140.
RDEB-O	103	p.Gly2569Arg	c.7705G>C	103	c.7705G>C	GS	p.Gly2569Arg	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
RDEB-O	103	p.Gly2569Ser	c.7705G>A	6	c.846+1G>A	SS	predicted skipping of exon 6 (in-frame; 55aa)	Kern et al. Br J Dermatol 2009; 161: 1089–1097
RDEB-O	103	p.Gly2576Arg[k]	c.7723G>A	116	c.8569G>T	PTC	p.Glu2857Stop	Shimizu et al. J Invest Dermatol 1996; 106: 119–124
RDEB-sev gen	104	p.Gly2587Asp	c.7760G>A	80	c.6527insC	PTC	p.Prol2176fsX113	Escámez MJ et al. Br J Dermatol 2010; 163: 155–161.
RDEB-O	104	p.Gly2590Asp	c.7769G>A	33	c.4011G>A	SS	p.Pro1337Pro	Van den Akker et al. J Dermatol Sci 2009; 56: 9–18.
RDEB-i	105	p.Gly2602Glu	c.7805G>A	68	c.5720G>A	GS	p.Gly1907Glu	Van den Akker et al. J Dermatol Sci 2009; 56: 9–18.
RDEB-O	105	**p.Gly2620Val**	c.7859G>T	105	c.7864C>T	MS	p.Arg2622Trp	**This paper**
RDEB-O	105	p.Gly2623Ser	c.7867G>A	70	c.5818delC	PTC	p.Pro1940fsX64	Sawamura et al. Br J Dermatol 2006; 155: 834–837.

RDEB-O	107	p.Gly2653Arg	c.7957G>A	97	c.7411C>T	PTC	p.Arg2471Stop	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
RDEB-pr	107	**p.Gly2656Val**	c.7967G>T	65	c.5564ins28	PTC	p.Arg1855fsX34	**This paper**
RDEB-sev gen	108	p.Gly2671Val	c.8012G>T	108	c.8012G>T	GS	p.Gly2671Val	Kon et al. J Invest Dermatol 1997; 108: 224–228.
RDEB-sev gen	108	p.Gly2674Arg	c.8020G>C	79	c.6501G>A	SS	in-frame deletion	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
RDEB-O	108	p.Gly2674Asp	c.8021G>A	13	c.1732C>T	PTC	p.Arg578Stop	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-O	109	p.Gly2689Arg	c.8065G>A	80	c.6527insC	PTC	p.Prol2176fsX113	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB	109	p.Gly2695Ser	c.8083G>A	109	c.8083G>A	GS	p.Gly2695Ser	Varki et al. J Med Genet 2007; 44: 181–192.
RDEB-sev gen	110	p.Gly2719Ala	c.8156G>C	ND	ND	ND	ND	Fassihi et al. J Dermatol Sci 2006; 42: 241–248.
RDEB	110	p.Gly2737Arg	c.8209G>C	ND	ND	ND	ND	Kern et al. J Invest Dermatol 2006; 126: 1006–1012.
RDEB-sev gen	110	p.Gly2740Ala	c.8219G>C	104	c.7786delG	PTC	p.Gly2596fsX49	Whittock et al. J Invest Dermatol 1999; 113: 673–686.
RDEB-sev gen	111	p.Gly2749Arg	c.8245G>A	111	c.8245G>A	GS	p.Gly2749Arg	Christiano et al. Am J Hum Genet 1996; 58: 671–681.
RDEB-O	112	p.Gly2775Ser	c.8323G>A	3	c.425A>G	SS[c]	p.Lys142Arg	Kon et al. J Invest Dermatol 1998; 111: 534–537.
RDEB-BDN	112	**p.Gly2781Arg**	c.8341G>A	51	c.4894C>T	PTC	p.Arg1632Stop	**This paper**

RDEB: recessive dystrophic epidermolysis bullosa; RDEB-sev gen: severe generalized; RDEB-O: other; RDEB-BDN: bullous dermolysis of the newborn; RDEB-na: nails only; RDEB-pr: pruriginosa; RDEB-i: inversa; GS: glycine substitution; MS: missense; N: normal; PTC: premature termination codon; ND: not determined; NR: not recorded; SS: splice site; **bold**: new mutations.

[a]within donor splice site consensus sequence, [b]results in a premature termination codon; [c]results in the loss of a StyI restriction site with various mRNA isoforms (Gardella et al. Am J Hum Genet 1996; 59: 292–300); [e]within acceptor splice site consensus sequence; corrections: [d]p.Gly1696Glu, [f]p.Gly1907Asp, [g]c.6080G>A, [j]p.Gly2375Ser, [k]p.Gly2576Arg, but corrected nomenclature should be: p.Gly1697Glu, p.Gly1908Asp, c.6091G>A, p.Gly2377Ser, p.Gly2575Arg, respectively; [h]*De novo* mutations; [i]inherited on the same allele.