Table SI. *Clinical and molecular findings in patients with bullous dermolysis of the newborn*

				Disease extent				
Reference	Sex	Inheri-tance	Congenital skin defects	Skin	Oral mucosa	Nails	Age at skin fragility resolution	*COL7A1* mutations[a]
Hashimoto et al. (1)	M	NK	No	Generalized	No	No	12 months	NK
Hashimoto et al. (9)	M	NK	No	Generalized	No	No	17 months	NK
	F	NK	No	Generalized	No	No	6 weeks	NK
Fine et al. (5)	M	AD[b]	No	Generalized	NR	No	3 months	NK
	M	NK	No	Generalized	NR	Yes	NK (mild disease activity at 1.5 years)	NK
Fine et al. (5); Fine et al. (10)	M	NK	No	Generalized	NR	Yes	6 months	NK
	M	AD[b]*	No	Generalized	NR	No	3 months	NK
Fine et al. (10)	NR	AD[b]*	NR	NR	NR	NR	3 months	NK
	F	AD[b]**	No	Generalized	Yes	Yes	NK (mild disease activity at 6 months)	NK
	F	AD[b]**	No	Generalized	NR	Yes	6 months	NK
	M	AD***	No	Generalized	Yes	Yes	NK (residual disease activity at 13 months)	c.4120-1G>C (heterozygous)[c]
	M	AD***	No	NK	NK	Yes	NK (no blisters at 23 years)	c.4120-1G>C (heterozygous)[c]
	F	AD***	No	NK	NK	NR	NK (mild disease activity at 11 years)	c.4120-1G>C (heterozygous)[c]
	F	AD***	No	NK	NK	Yes	NK (no blisters at 37 years)	c.4120-1G>C (heterozygous)[c]
Smith & Sybert (6)	M	AR***	No	Generalized	Yes	NR	NA (died at 10 days, cardio-pulmonary arrest)	NK
McCollough et al. (11)	F	AD[b]	No	Generalized	Yes	Yes	NK (mild disease activity at 6 months)	NK
Eng et al. (12)	F	NK	No	Generalized	No	No	4 weeks	NK
Patrizi et al. (13)	F	AR[b]	Yes (lower arms and feet)	Generalized	No	No	18 months	NK
Phillips et al. (14)	F	NK	No	Generalized	NR	Yes	NK (mild disease activity at 6 months)	NK
Okuda et al. (15)	F	NK	No	Generalized (a few lesions)	No	No	1 month	NK
Hatta et al. (16)	F	NK	Yes (right leg and knee and feet)	Generalized	Yes	Yes	NK (residual disease activity at 4 years)	NK
D'incan et al. (17)	F	NK	No	Generalized	No	No	3 weeks	NK
Hammami-Hauasli et al. (4)	F	AR	Yes (lower legs)	Generalized	NR	Yes	NK (mild disease activity at 14 months)	p.Gly1519Asp/p.Gly2251Glu
Hanson et al. (18)	M	NK	No	Localized (acral)	NR	NR	1 month	NK
	F	NK	Yes (from knees to toes)	Generalized	Yes	NR	NK (residual disease activity at 7 months)	NK
	M	NK	No	Localized (acral)	Y	NR	2 months	NK
Fassihi et al. (19)	M	AD	No	Localized (acral)	No	No	4 months	p.Gly1522Glu (heterozygous)
Nakano et al. (20)	M	AR	Yes (first toes)	Generalized	Yes	Yes	29 months	p.Arg990Gln in linkage with c.5504delA/p.Arg2008His
Oh et al. (21)	F	AR	Yes (right lower leg)	Localized (acral)	NR	NR	2 months	p.Gly798Arg/c.6246del27
Hashikawa et al. (22)	M	AR	No	Generalized	NR	Yes	12 months	c.682+1G>A/p.Gly1910Ser
Oppenheimer & Hallas (23)	F	AR[b]	No	Generalized	No	NR	NK (residual disease activity at 3 months)	NK
Almaani et al. (24)	F	AD	No	Generalized	NR	NR	4 months	p.Gly1483Asp (heterozygous)
Frew et al. (25)	M	AD	No	Generalized	No	NR	18 months	p.Gly1673Arg (heterozygous)
Murase et al. (26)	M	AD	No	Localized (acral)	No	No	6 months	p.Gly2242Glu (heterozygous)
Radkevich-Brown &	M	AR[b]	Yes (right foot)	Generalized	Yes	Yes	7 months	NK
Schwayder (27)	M	AR[b]	Yes (left foot)	Localized (acral)	Yes	Yes	4 months	NK
	M	NK	No	Generalized	NR	NR	3 months	NK
	M	NK	No	Generalized	Yes	Yes	5 months	NK
Boccaletti et al. (28)	F	AR	No	Generalized	Yes	Yes	NK (mild disease activity at 3 years)	p.Pro2259Leu (homozygous)
	M	AR	No	Generalized	Yes	Yes	NK (minimal disease activity at 10 months)	p.Pro2259Leu (homozygous)
Shi et al. (29)	M	AD	Yes (pretibial areas)	Localized (legs, feet)	No	No	2 months	p.Gly2046Ser
Current study								
1	F	AR	Yes (from knees to feet)	Generalized	Yes	Yes	3 months	c.497dupA/p.Gly2216Glu
2	M	AR	Yes (left leg, from knee to foot)	Localized (lower extremities and face)	Yes	No	3 months	c.4783-1G>A/ p.Pro1699Leu
3	M	AD	No	Localized (extremities and face)	No	Yes	Residual disease activity at 3 years	p.Gly2431Val
4	M	AD	No	Generalized	No	Yes	3 months	p.Gly1830Arg

At least 4 additional molecularly characterized cases were reported, but were not included in the analysis because of insufficient clinical description (24, 30).
[a]Mutation designation is based on the cDNA sequence deposited under the GenBank entry NM_000094.3. [b]Inheritance based on suggestive family history. [c]The mutation in this family has been identified and described by Christiano et al. (3). *Members of family 2 (10); **members of family 3 (10); ***members of family 4 (10).
NR: not reported; NK: not known; NA: not applicable.