

Table S1. Details of the mutations of all the Australian epidermolysis bullosa simplex (EBS) patients involved in our study

Family No.	Pat. No.	EBS subtype	Gene	Mutation (c.)	Mutation (p.)	Domain	Phenotype in previous reports
EB294	543	Gen. intermed.	KRT5	280G>A, 434T>G	G94S V145G	Head	Novel
	544	Gen. intermed.	KRT5	280G>A, 434T>G	G94S V145G	Head	Novel
	294	Gen. intermed.	KRT5	280G>A, 434T>G	G94S V145G	Head	Novel
	520	Gen. intermed.	KRT5	280G>A, 434T>G	G94S V145G	Head	Novel
	386	Gen. intermed.	KRT5	280G>A, 434T>G	G94S V145G	Head	Novel
EB411	411	Gen. severe	KRT5	449T>C	L150P	Head	Gen. intermed. (Kang et al., 2010; Oh, Lee, Kim, & Kim, 2007); Gen. severe (Bchethnia et al., 2012; Minakawa et al., 2013)
EB440	440	Localised	KRT5	482T>G	I161S	Head	Localised (Y. M. Chan, Yu, Fine, & Fuchs, 1993; Ehrlich, Sybert, Spencer, & Stephens, 1995; Pfendner et al., 2003)
EB330	519	Localised	KRT5	482T>G	I161S	Head	Novel
	339	Localised	KRT5	502G>C	E168Q	Head	Novel
	330	Localised	KRT5	502G>C	E168Q	Head	Novel
	567	Localised	KRT5	502G>C	E168Q	Head	Novel
	70	Gen. severe	KRT5	514A>T	I172F	1A	Novel
EB30	8	Gen. intermed.	KRT5	519G>C	K173N	1A	Gen. intermed. (Stephens et al., 1995)
	273	Gen. intermed.	KRT5	519G>C	K173N	1A	Gen. intermed. (Stephens et al., 1995)
EB534	30	Gen. intermed.	KRT5	519G>C	K173N	1A	Gen. intermed. (Stephens et al., 1995)
	553	Localised	KRT5	535T>C	F179L	1A	Novel
EB74	534	Localised	KRT5	535T>C	F179L	1A	Novel
	74	Gen. intermed.	KRT5	548T>A	I183N	1A	Novel
EB344	344	Localised	KRT5	968T>C	V323A	L12	Gen. intermed. (Galligan et al., 1998)
EB459	459	Localised	KRT5	968T>C	V323A	L12	Gen. intermed. (Galligan et al., 1998)
	460	Localised	KRT5	968T>C	V323A	L12	Gen. intermed. (Galligan et al., 1998)
EB28	28	Gen. severe	KRT5	1285_1299del15	DELTA RNKLA 429-433	2B	Gen. severe (Kemp et al., 2005)
	331	Localised	KRT5	1401C>G	I467M	2B	Gen. intermed. (Bolling et al., 2011; Pfendner et al., 2005)
EB35	289	Localised	KRT5	1401C>G	I467M	2B	Gen. intermed. (Bolling et al., 2011; Pfendner et al., 2005)
	300	Localised	KRT5	1401C>G	I467M	2B	Gen. intermed. (Bolling et al., 2011; Pfendner et al., 2005)
EB286	332	Localised	KRT5	1401C>G	I467M	2B	Gen. intermed. (Bolling et al., 2011; Pfendner et al., 2005)
	286	Gen. intermed.	KRT5	1427G>C	G476D	2B	Localised (Abu et al., 2006; Bolling, Lemmink, Jansen, & Jonkman, 2011; Kowalewski et al., 2007)
EB256	256	Gen. severe	KRT5	1429G>A	E477K	2B	Gen. severe (Arim et al., 2010; Garcia et al., 2011; Jerabkova et al., 2010; Kang, Lee, Kim, Oh, & Kim, 2010; Muller et al., 2006; Pfendner, Nakano, Pulkkinen, Christiano, & Utto, 2003; Pfendner, Sadowski, & Utto, 2005; Stephens et al., 1997)
	266	Gen. severe	KRT5	1429G>A	E477K	2B	Gen. severe (Arim et al., 2010; Garcia et al., 2011; Jerabkova et al., 2010; Kang, Lee, Kim, Oh, & Kim, 2010; Muller et al., 2006; Pfendner, Nakano, Pulkkinen, Christiano, & Utto, 2003; Pfendner, Sadowski, & Utto, 2005; Stephens et al., 1997)
EB26	26	Gen. severe	KRT14, KRT5	1237G>A, 504G>C	A413T, E168D	2B, 1A	Gen. intermed. (Shinkuma et al., 2013)
	557	Gen. severe	KRT14, KRT5	1237G>A, 504G>C	A413T, E168D	2B, 1A	Gen. intermed. (Shinkuma et al., 2013)
	27	Gen. severe	KRT14, KRT5	1237G>A, 504G>C	A413T, E168D	2B, 1A	Gen. intermed. (Shinkuma et al., 2013)
	233	Gen. intermed.	KRT14, KRT5	1237G>A, 597G>C	A413T K199N variant	2B, 1A	Gen. intermed. (Deng et al., 2011)
	361	Localised	KRT14	881T>C	M294T	2A	Novel (Digenic)
EB54	92	Gen. intermed.	KRT5	464T>C	L155P	Head	Gen. intermed. (Cummins et al., 2001), Localised (Yasukawa et al., 2006)
	57	Gen. intermed.	KRT14	355A>G	M119V	1A	Gen. intermed. (Cummins et al., 2001), Localised (Yasukawa et al., 2006)
	54	Gen. intermed.	KRT14	355A>G	M119V	1A	Gen. intermed. (Cummins et al., 2001), Localised (Yasukawa et al., 2006)
	93	Gen. intermed.	KRT14	355A>G	M119V	1A	Gen. intermed. (Cummins et al., 2001), Localised (Yasukawa et al., 2006)
	204	Gen. severe	KRT14	356T>C	M119T	1A	Gen. severe (Cummins et al., 2001; Jerabkova et al., 2010; Shemanko et al., 2000; Titeux et al., 2006), Mottled Pigmentation (Harel, Bergman, Indelman, & Sprecher, 2006)
EB394	394	Gen. severe	KRT14	365T>C	L122P	1A	Novel
	307	Gen. severe	KRT14	368A>T	N123I	1A	Novel
	37	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulombe et al., 1991; Hachisuka et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB289	289	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulombe et al., 1991; Hachisuka et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB312	312	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulombe et al., 1991; Hachisuka et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)

EB420	420	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulobme et al., 1991; Hachisuika et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB63	63	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulobme et al., 1991; Hachisuika et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB87	64	Gen. severe	KRT14	373C>T	R125C	1A	Gen. severe (Chen et al., 1995; Coulobme et al., 1991; Hachisuika et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB282	282	Gen. severe	KRT14	374G>A	R125H	1A	Gen. severe (Chen et al., 1995; Coulobme et al., 1991; Hachisuika et al., 1995; Hut et al., 2000; Ning et al., 2001; Pfendner et al., 2003; Premaratne et al., 2002; Rugg et al., 2000; Sasaki et al., 1999; Stephens et al., 1993; Umeki et al., 1996)
EB323	285	Gen. severe	KRT14	374G>A	R125H	1A	Gen. severe (Arim et al., 2010; Ciobotaru et al., 2003; Coulobme et al., 1991; Frangu et al., 2006; Horn & Tidman, 2000; Hut et al., 2000; Jerabkova et al., 2010; Kang et al., 2010; Muller et al., 2006; Rugg et al., 2007; Shemanko et al., 2000; Stephens et al., 1993; Wu & Xiao, 2009; Yasukawa et al., 2006), Localised (Minakawa et al., 2013)
EB516	283	Gen. severe	KRT14	374G>A	R125H	1A	Gen. severe (Arim et al., 2010; Ciobotaru et al., 2003; Coulobme et al., 1991; Frangu et al., 2006; Horn & Tidman, 2000; Hut et al., 2000; Jerabkova et al., 2010; Kang et al., 2010; Muller et al., 2006; Rugg et al., 2007; Shemanko et al., 2000; Stephens et al., 1993; Wu & Xiao, 2009; Yasukawa et al., 2006), Localised (Minakawa et al., 2013)
EB555	323	Gen. severe	KRT14	374G>A	R125H	1A	Gen. severe (Arim et al., 2010; Ciobotaru et al., 2003; Coulobme et al., 1991; Frangu et al., 2006; Horn & Tidman, 2000; Hut et al., 2000; Jerabkova et al., 2010; Kang et al., 2010; Muller et al., 2006; Rugg et al., 2007; Shemanko et al., 2000; Stephens et al., 1993; Wu & Xiao, 2009; Yasukawa et al., 2006), Localised (Minakawa et al., 2013)
EB572	516	Localised	KRT14	385T>C	Y129H	1A	Novel
EB91	555	Gen. severe	KRT14	442C>T	R148C	1A	Localised (Wood, Batty, Lane, & McLean, 2003)
EB274	72	Recessive Gen. intermed.	KRT14	612T>A	Y204X	1B	Recessive Gen. intermed. (Y. Chan et al., 1994; Ylaseimides et al., 2008)
15/y	91	Localised	KRT14	815T>C	M272T	L12	Gen. intermed. (Arim et al., 2010; Muller et al., 2006), Localised (Arim et al., 2010)
260/y	272	Localised	KRT14	815T>C	M272T	L12	Gen. intermed. (Arim et al., 2010; Muller et al., 2006), Localised (Arim et al., 2010)
281/y	279	Localised	KRT14	815T>C	M272T	L12	Gen. intermed. (Arim et al., 2010; Muller et al., 2006), Localised (Arim et al., 2010)
298/y	94	Localised	KRT14	815T>C	M272T	L12	Gen. intermed. (Arim et al., 2010; Muller et al., 2006), Localised (Arim et al., 2010)
368/y	280	Localised	KRT14	815T>C	M272T	L12	Gen. intermed. (Arim et al., 2010; Muller et al., 2006), Localised (Arim et al., 2010)
372/h	46	Gen. intermed.	KRT14	863G>T	R288L	2A	Gen. intermed. (Minakawa et al., 2013)
80/n	45	Gen. intermed.	KRT14	863G>T	R288L	2A	Gen. intermed. (Minakawa et al., 2013)
95/n	422	Localised	KRT14	1237G>A	A413T	2B	Gen. intermed. (Chao et al., 2002; Minakawa et al., 2013)
68/n	423	Localised	KRT14	1237G>A	A413T	2B	Gen. intermed. (Chao et al., 2002; Minakawa et al., 2013)
	239	Localised	KRT14	1237G>A	A413T	2B	Gen. intermed. (Chao et al., 2002; Minakawa et al., 2013)
	89	Localised	KRT14	1253T>A	L418Q	2B	Localised & Gen. intermed. (Wertheim-Tysarowska et al., 2015)
	503	Gen. severe	KRT14, KRT14	1250_1252 del GCC, 1237G>A	R417del, A413T	2B, 2B	Novel
EB314	314	Localised					No mutations detected
EB378	391	Localised					No mutations detected
EB20	378	Localised					No mutations detected
EB274	20	Localised					No mutations detected
15/y	276	Localised					No mutations detected
260/y	274	Localised					No mutations detected
281/y	15	Localised					No mutations detected
298/y	260	Localised					No mutations detected
368/y	281	Gen. intermed.					No mutations detected
372/h	298	Localised					No mutations detected
80/n	368	Localised					No mutations detected
95/n	372	Localised					No mutations detected
68/n	80	Gen. intermed.					No mutations detected
	95	Gen. severe					No mutations detected
	68	Gen. severe					No mutations detected

*A413T may or may not cause disease (Murrell, Trisnowati, Miyakis, & Paller, 2011).