

**Table SII. Genotype spectrum of patients with recessive dystrophic epidermolysis bullosa from India**

Family	Patient	Total variants in 18 genes	COL7A1 cDNA change	Exon	Protein Change	SIFT	PolyPhen	Consequence	Status	Reference
7	8	251	c.172dupC/ c.172dupC	2	R58fs/R58fs	NA/NA	NA/NA	PTC/PTC	Novel/Novel	-
8	9	162	c.1893G>A/ c.1893G>A	14	W631X/W631X	NA/NA	NA/NA	PTC/PTC	Reported/ Reported	Christiano et al, 1996 (19)
9	10	342	c.3832-2A>G/ c.3832-2A>G	Intron 31- Exon 32	-/-	NA/NA	NA/NA	PTC/PTC	Reported/ Reported	Kern et al, 2006 (20)
10	11	249	c.8047-1G>A/ c.8047-1G>A	Intron 109- Exon 110	-/-	NA/NA	NA/NA	PTC/PTC	Novel/Novel	-
11	12	128	c.3054delA/ c.3310_3311insA	23/25	L1018fs/P1104fs	NA/NA	NA/NA	PTC/PTC	Novel/Novel	-
12	13	290	c.6073G>A/ c.6073G>A	73	G2025S/G2025S	0.01 (D)/0.01 (D)	1 (D)/1 (D)	Missense/ Missense	Reported/ Reported	Pfender et al, 2003 (21)
13	14	247	c.520G>A/ c.3850G>A	4/31	G174R/G1284S	NA/0.04 (D)	1 (D)/1 (D)	Missense/ Missense	Reported/ Novel	Kern et al, 2009 (24)
14	15	388	c.4894C>T/ c.6007G>A	51/73	R1632X/G2003R	NA/0 (D)	NA/1 (D)	PTC/Missense	Reported/ Reported	Whitlock et al, 1999 (27)/Christiano et al, 1996 (19)
15	16	128	c.409C>T/ c.8676G>A	3/117	R137X/W2892X	NA/NA	NA/NA	PTC/PTC	Reported/ Novel	Sawamura et al, 2005 (28)
16	17	164	c.448G>A/ c.3310_3311insA	4/25	G150R/P1104fs	0.03 (D)/NA	1 (D)/NA	Missense/PTC	Reported/ Novel	Drera et al, 2009 (26)-
17	18	123	c.520G>A/ c.5487+1G>-	4/Intron 63-Exon 64	G174R/-	NA/NA	1 (D)/NA	Missense/PTC	Reported/ Novel	Kern et al, 2009 (24)

NA: not available; D: deleterious; PTC: premature termination codon.