Table SI. Phenotypic spectrum of patients with recessive dystrophic epidermolysis bullosa from India

Family	Patient	Age/Sex	Phenotype	Disease onset	Consan-guinity	IFM (COLVII) (Normal healthy control: No split/ +++)	Distribution	Scarring	Milia	Nail	Mucosal	Eye	Additional features
7	8	2 years/M	RDEB_GS	Birth	No	Sub epidermal split/ Absent	Generalized	++	++	++	++	++	Microstomia, syndactyly, alopecia
8	9	2 years/M	RDEB_GS	Birth	Yes	Sub epidermal split/ Absent	Generalized	++	++	++	++	+	Microstomia, syndactyly
9	10	6 months/M	RDEB_GS	Birth	No	Sub epidermal split/ Absent	Generalized	++	++	++/+	++	+	Syndactyly
10	11	6 months/F	RDEB_GS	Birth	No	Sub epidermal split/ Absent	Generalized	+	++	++/+	+	+	NA
11	12	2 years/M	RDEB_GS	Birth	No	Sub epidermal split/ Absent	Generalized	++	+	++/+	+	+	NA
12	13	24 years/M	RDEB_GI	Birth	No	Sub epidermal split/+/–	Generalized	++	++	++	++	+	Microstomia, alopecia
13	14	4 years/F	RDEB_GI	Birth	No	Sub epidermal split/ absent	Generalized	++	++	++	+	+	NA
14	15	3 years/F	RDEB_GI	Birth	No	Sub epidermal split/+/–	Generalized	++	++	++	++	+	Microstomia, syndactyly, alopecia
15	16	14 years/M	RDEB_GI	Birth	No	Focal cytolysis; ++	Generalized	+	+	++/+	+	–	NA
16	17	5 years/F	RDEB_GI	Birth	Yes	Focal cytolysis/+	Generalized	++	++	++	+	+	NA
17	18	12 years/M	RDEB_GI	6 months	No	Focal cytolysis; ++	Limited	+	+	+	+	–	NA

IFM: immunofluorescence antigen mapping; GS: generalized severe; GI: generalized intermediate; RDEB: recessive dystrophic epidermolysis bullosa.