

Supplement Data

Table S1. The summary of genotype-phenotype correlation of *COL7A* variants identified in family 3-4 based on previous reports

Gene	Variant 1	Variant 2	Inheritance	Age/gender	Skin / mucosal blistering	Pseudosyndactyly	Dysphagia	Nail dystrophy	Dental abnormalities	Growth retardation	Symblepharon	Reference
COL7A1	c.6187C>T p. R2063W	c.344dupG	compound	14-year-old	General,	Yes	Yes	NA	NA	Yes	NA	[19]
		p. N116fs	heterozygous	male	Lethal							
	c.7097G>A p. G2366D	c.4965C>T	compound	25-year-old	Yes	No	Yes	Yes	NA	Yes	NA	[19]
			heterozygous	female								
	c.3625_3635del p. S1209fs	c.6527dupC	compound	NA	Yes	No	Yes	Yes	NA	NA	Yes	[23]
		p. G2177fs	heterozygous									
	c.8569G>T p. E2857*	c.142T>C	compound	NA	General,	Yes	Yes	Yes	NA	NA	NA	[20]
		p. S48P	heterozygous	male	Lethal							
	c.8569G>T p. E2857*	c.6573+1G>A	compound	3-year-old	Yes	No	No	Yes	No	NA	NA	[21]
			heterozygous	male								
	c.8569G>T p. E2857*	c.2005C>T	compound	19-year-old	Yes	No	Yes	Yes	Yes	NA	NA	[22]
		p.R669*	heterozygous	female								

Variant 1: the reported variant identified in our study;

Variant 2: another compound heterozygous variant in the previous reports.