

Table SI. Molecular and clinical features of the patients

Cases, age/clinical features	Case 1 Male, age 11 years	Case 2 Female, age 11 years	Case 3 Female, age 6 years	Case 4 Male, age 5 years
Molecular features				
COL17A1 mutations	c.[2407G>T];[3198C>T] p.[G803*];[S1066S]	c.[2407G>T];[3198C>T] p.[G803*];[S1066S]	c.[2237delG];[3198C>T] p.[G746Afs*53];[S1066S]	c.[3198C>T];[4156+1G>A] p.[S1066S];[?]
Relative collagen XVII expression by IFM	Strongly reduced	Strongly reduced	Reduced	Reduced
Age at last examination	10 years	3.5 months	1 month	5 years
Skin	Blisters, erosions, atrophy, hypopigmentation	Skin defects, blisters	Blisters, erosions	Blisters, erosions, scarring
Palmoplantar skin	Blisters, erosions, plantar hyperhidrosis	Blisters	None	Blisters, erosions
Hair	Not affected	Not affected	Not affected	Not affected
Nails	Dystrophic, onycholysis, nail-loss	Dystrophic, nail-loss	Dystrophic, nail-loss	Dystrophic
Mucosal involvement	Oral, ocular pain due to corneal erosions, genitourinary	Oral, nasal, genital	Nasal, oral	Oral, nasal, ocular pain due to corneal erosions
Teeth	Caries, enamel defects	Not available	Not available	Enamel defects
Parental dental status	Enamel defects (pits), no caries	Parents none	Mother (het. 3198C>T) periodontitis, caries Father (het. 2237delG) none	Caries