

Table SI. Clinical and genetic data in reported cases of segmental Darier disease

Case (ref.)	Sex	Sporadic/familial	Age/age at onset (years)	Disease manifestations	ATP2A2 mutation (cDNA, protein)	Tissues analysed for mutation search
1 (6)	F	S	48/11	Warty papules in a linear pattern on the right leg, trunk, arm and forehead, following Blaschko's lines	c.2682C>A, p.Tyr894* (exon 18)	AE: + NIE: - B: -
2 (6)	M	S	35/17	Papules on the right trunk and back, then right inner thigh following Blaschko's lines	c.2305G>A, p.Gly769Arg (exon 15)	AE: + NIE: - B: -
3 (11)	F	S	41/38	Hyperkeratotic papules on the left side of the trunk following Blaschko's lines	c.160A>G, p.Ile54Val (exon 3)	AE: + NIE: - B: -
4 (4)	M	S	29/12	Hyperkeratotic papules on the lower extremities; then asymmetric involvement of scalp, face, ears, back, left arm, groin, neck and chest (on the back, left arm and lower extremities lesions followed Blaschko's lines); palmar pits; longitudinal leukonychia of fingernails	c.68G>A, p.Gly23Glu (exon 1)	AE: + (37%)* NIE: + (21%)* B: + (24%)* S: + (14%)* H: + (35%)*
5 (8)	M	Fam	1/shortly after birth	Erosive plaques on the right side of trunk and abdomen (diagnosed as segmental DD type 2)	c.1030G>A, p.Gly343Asp (exon 8)	AE: + B: +
6 (9)	F	S	62/ 57	Bilateral papules on mons pubis, external labia, inguinal folds, upper medial thighs, central chest, inframammary folds, not arranged in a Blaschkoid pattern	c.2116G>A, p.Asp706Asn (exon 15)	AE: + NIE: - B: -
Present case	F	S	47/27	Hyperkeratotic papules on the right aspect of the chest, right arm and right lower limb	c.2256_2258delCTA, p.Tyr753del (exon 15)	AE: + NIE: - B: -

Fam: familial; S: sporadic; AE: affected epidermis; NIE: non-involved epidermis; B: blood; S: semen; H: hair; + present; - not present; DD: Darier disease; *Percentage of mutated alleles found in the tissue examined.