Table S1: Desmosomal disorders related to *JUP* mutation(s)

Disease	Dermatological symptoms				Cardiac involvement	Mode of inheritance	Mutation	Location	Total of patients	Total of families	Origin
	PPK	Hair	Skin fragility	Others ectodermal features							
Naxos syndrome	+	woolly +/- sparse	-	-	ARVC	recessive	c.2157del2	Exon 14	74	NA	Greek / Turkish [1–5]
CAPK syndrome	+	atrichia	-	-	ARVC	recessive	c.794G>A/ p.R265H	Exon 4	2	1	Turkish [6]
Skin fragility + PPK + Woolly hair	+	woolly + sparse	+	-	no	recessive	c.71C>A/ p.S24X	Exon 2	3	1	Argentine [7]
							c.468G>A	Exon 3	2	1	Kuwaiti [7]
Lethal congenital epidermolysis bullosa	-	alopecia	+	onycholysis	no	recessive	c.1615C>T/ p.Q539X	Exon 9	1	1	German [8]

Sequences variations of *JUP* are referenced in the transcript GENEBANK accession NM_002230.

Table S2: Desmosomal disorders related to *DSP* mutation(s)

Disease	Dermatological symptoms				Cardiac involve-ment	Mode of inheritance	Mutation	Location	Total of patients	Total of families	Origin
	PPK	Hair	Skin fragility	Others ectodermal features							
Striate PPK	+	-	-	-	no	dominant	c.939+1G>A	Intron 7	3	1	NA [9]
							c.C991T/p.Q331X*	Exon 8	27	1	NA [10]
Autosomal dominant Carvajal Syndrome	+	woolly and/or sparse	-	oligodontia	yes	dominant	c.1691C>T/p.T564I	Exon 13	3	2	NA [11,12]
				oligodontia			30bp insertion	Exon 14	2	1	NA [13]
				oligodontia leuconychia			c.1790C>T/p.S597L	Exon 14	3	1	NA [14]
				nail dystrophy			c.1748T>C/p.L583P	Exon 14	1	1	Caucasian [12]
Autosomal recessive Caravajal Syndrome	+	woolly and/or sparse	-	-	yes	recessive	c.7901delG	Exon 24	12	3	Ecuadorian [15]
							c.3799C>T/p.R1267X	Exon 23	1	1	Turkish [16]
							c.6166G>C/p.G2056R	Exon 24	1	1	Danish [17]
							c.3901C>T/p.Q1301X	Exon 23	1	1	Indian [18]
							c.5208_5209delAG/ p.G1737TfsX7	Exon 24	2	1	NA [19]
							c.7780delT /p.S2594PfsX8	Exon 24	1	1	Turkish [20]
							c.7567delAAGA/p.K2523fsX37 c.6577G>A/p.E2193K	Exon 24 Exon 24	1	1	NA [21]

							c.7566_7567delAAinsC/ p.R2522SfsX39 c.7756C>T/ p.R2586X	Exon 24 Exon 24	1	1	NA [22]
							c.1067C>A/ p.T356K c.2131_2132delAG/ p.S711CfsX4	Exon 9 Exon 16	2	1	NA [22]
							c.7123G>C/p.G2375R	Exon 24	9	1	Israeli [23]
							c.3924delG/p. H1309TfsX1348	Exon 23	1	1	Palestinian [24]
							c.7111C>A, Q2371K	Exon 24	5	1	Palestinian [24]
Carvajal and skin fragility syndrome	+	woolly and/or sparse	+	-	yes	recessive	c.4336C>T/p.Q1446X c.2017C>T/p.Q673X	Exon 23 Exon 15	1	1	UK [25]
				nail dystrophy			c.4778_4790del13/p. L1593KfsX5 c.6310delA/p.T2104QfsX12	Exon 23 Exon 24	1	1	Swedish [26]
				nail dystrophy			c.4198C>T/R1400X c.6850C>T/R2284X	Exon 23 Exon 24	1	1	Dominican [27]
				teeth abnormalities nail dystrophy			c.6310delA/ p.T2104QfsX12 c.7964C>A/ p.A2655D	Exon 24 Exon 24	1	1	Finnish [28]
				nail dystrophy			c.2516del4/p.H839fsX23 c.3971del4/p.N1324f	Exon 18 Exon 23	1	1	Brazilian [29]

							sX23				
Skin fragility and woolly hair syndrome	+	woolly and/or sparse	+	nail dystrophy	no	recessive	c.2427T>A/p.C809X c.861G>T/p.N287K	Exon17 Exon 7	1	1	NA [30]
							c.1990C>T/p.Q664X c.7096C>T/p.R2366C	Exon 15 Exon 24	1	1	NA [30]
							c.7097G>A/ p.R2366H	Exon 24	5	1	Saudi Arabian [31]
							c.7096C>T/p.R2366C c.6721_6722delAT/p.I2241FfsX3	Exon 24 Exon 24	1	1	Caucasian [32]
Lethal acantholytic epidermolysis bullosa		alope -cia	+	anonychia	poor cardiac contracti -lity in one patient	recessive	c.5800C>T/p.R1934X ** c.6370delTT	Exon 24 Exon 24	1	1	Dutch [33]
							c.2874del5/p.L959MfsX5	Exon-In -tron 20	2	1	Yemeni [34]
							c.7248delT/p.F2416LfsX14	Exon 24	1	1	NA [35]

Sequences variations of *DSP* are referenced in the transcript GENEBANK accession NM_004415.
c.C991T/p.Q331X* : this mutation was published as c.C1323T/p.Q331X. The correspondence between c.C1323T and p.Q331X was not found in ENSEMBL software. Therefore, we renamed it with this new combination.
c.5800C>T/p.R1934X** : This mutation was published as c.C6079T/p.R1934X . The correspondence between c.C6079T and p.R1934T was not found in ENSEMBL software. Therefore, we renamed it with this new combination.

Table S3: McGrath Syndrome, *PKP1* identified mutations

Disease	Mode of inheritance	Mutation	Location	Total of patients	Origin
: Ectodermal Dysplasia/Skin fragility syndrome : - PPK - Skin fragility - Perioral lesions (at least 11 patients) - Sparse hair - Nail dystrophy - No cardiac involvement	recessive	c.203-1G>A c.213T>G/p.Y71X	Intron 1 Exon 2	1	Caucasian [36]
		c.203-1G>A	Intron 1	3	Arab [37]
		c.203-1G>T	Intron 1	2	Egyptian [38]
		c.847-2A>G	Intron 4	1	Arab [37]
		c.888delC	Exon 5	2	Turkish [39]
		c.897del5/p.P299fsX61	Exon 5	1	Iraqi [40]
		c.910C>T/p.Q304X c.1132ins28/PTC	Exon 5 Exon 6	1	British [41]
		c.1054+1G>T c.1835-2A>G	Intron 5 Intron 10	1	Chinese [42]
		c.1233+2A>T	Intron 6	1	French [43]
		c.1233-2A>G/p.R411SfsX51	Intron 6	1	Caucasian [44]
		IVS9+1G>A	Intron 9	1	Dutch [45]
		c.2014C>T / p.R672X	Exon 11	3	Brazilian [29]
		c.2021+1G>A	Intron 11	1	Japanese [46]

Sequences variations of *PKP1* are referenced in the transcript GENEBANK accession NM_001005337.

Table S4: Desmosomal disorders related to *DSG1* and *DSG4* mutation(s)

Disease	Gene	Mode of inheritance	Mutation	Location	Total of patients	Origin
Isolated PPK	***Desmoglein 1***	dominant	IVS2-1G>A	Intron 2	6	Dutch [47]
			c.76C>T/p.R26X	Exon 2	11	Yemenite/pakistani [48–50]
			c.121insT/PTC	Exon 3	3	Libyan [51]
			IVS4-2A>G	Intron 4	11	Pakistani [50]
			c.395C>A/p.S132X	Exon 5	3	Iranian-Syrian [52]
			c.430A>T/p.R144X	Exon 5	4	Scottish [53]
			c.515C>T	Exon 5	10	Pakistani [50]
			c.C602T/p.Q201X	Exon 5	5	Jewish Sephardic [54]
			c.C655T/p.R219X	Exon 5	3	Jewish Ashkenazi [54]
			c.1079insC/PTC	Exon 9	1	NA [48]
			c.1095T>A/p.Y365X	Exon 9	1	NA [48]
			c.1189delA/PTC	Exon 9	1	NA [48]
			IVS9-3C>G	Intron 9	10	Pakistani [50]
			c.1399delA	Exon 10	3	Pakistani [50]
			c.1627delA/PTC	Exon 11	1	NA [48]
			c.1668-1G>T	Intron 11	11	Pakistani [55]

			c.1931delA /PTC	Exon 14	2	Jewish Ashkenazi [54]
SAM Syndrome : - Severe **S**kin dermatitis, - multiple **A**llergies - **M**etabolic wasting	*Desmoglein 1*	recessive	c.49-1G>A, in-frame skipping of exon 2	Exon 2	2	Arab [56]
			c.1861delG/ p.A612Qfs.X3	Exon 13	4	Arab [56]
			c.2659C>T/p.R887X	Exon 15	1	NA [57]
Localized Autosomal recessive Hypotrichosis (LAH Syndrome)	*Desmoglein 4*	recessive	c.Ex5_8del	Exon 5 to 8	34	Pakistani [58–60]
			c.GG384-385TT/p.A129S	Exon 5	1	Iraqi [61]
			c.87delG/PTC	Exon 3	6	Pakistani [62]
Monilethrix	*Desmoglein 4*	recessive	c.574T>C/p.S192P c.2039insT/PTC	Exon 6 Exon 13	1	Japanese [63]
			c.799C>G/ P267R	Exon 7	9	Iraqi [64]
			p.P267R p.R289X	Exon 7 Exon 8	3	Iraqi / Maroccan [64]
			c.216+1G>T p.P267R	Intron 3 Exon 7	6	Iraqi and Iranian [64,65]
			c.763delT c.216+1G>T	Exon 7 Intron 3	2	Iranian [64]
			p.P267R c.763delT	Exon 7 Exon 7	2	Iraqi and Iranian [64]

	c.624delG/p.M208lfsX4 c.2468G>A/ p.W823X	Exon 6 Exon 16	1	Japanese [66]
	c.2119delG/p.R707IfsX109	Exon 14	1	Japanese [67]

Sequences variations of *DSG1 and DSG4* are referenced in the transcript GENEBANK accession NM_001942 and NM_ 177986 respectively.

Table S5: Desmosomal genodermatoses induced by *DSC2* and *DSC3* mutation(s)

Disease	Gene	Mode of inheritance	Mutation	Location	Total of patients	Origin
- Woolly hair - Mild PPK - Left cardiac involvement	*Desmocollin 2*	recessive	c.1841delG/ p.S614fsX625	Exon 12	2	NA [68]
Hypotrichosis	*Desmocollin 3*	recessive	c.2129T>G/p.L710X	Exon 14	4	Afghan [69]

Sequences variations of *DSC2 and DSC3* are referenced in the transcript GENEBANK accession NM_024422 and NM_001941, respectively.

Table S6: Desmosomal disorders related to *CDSN* mutation(s)

Disease	Mode of inheritance	Mutation	Location	Total of patients	Total of families	Origin
Hypotrichosis Simplex of the Scalp (HSS)	dominant	p.Q215X	Exon 2	26 (Israeli family) + 17 (spanish family)	2	Spanish and Israeli [70]
		p.Q200X	Exon 2	US	1	Danish [70]
		p.Y239X*	Exon 2	42	1	Mexican [71]
		c.717C>G/p.Y239X*	Exon 2	3	1	Chinese [72]
Peeling Skin Syndrome type B (PSS)	recessive	c.164_167dup GCCT/ p.T57PfsX6	Exon 2	1	1	Ashkenazi Jewish [73]
		c.175A>T/p.K59X	Exon 2	4	1	German [74]
		c.424G>T/p.G142X	Exon 2	2	1	NA [75]
		c.746delG/ p.G249VfsX40	Exon 2	2	1	NA[76]
		A genomic deletion at the PSORS1 locus removing the entire *CDSN* gene		1	1	Japanese [77]
		A large homozygous deletion of 59,184bp extending from 40.6 kb upstream to 13.2 kb downstream of *CDSN*		1	1	Japanese [78]

Sequences variations of *CDSN* are referenced in the transcript GENEBANK accession NM_001264.
* These two mutations seem to be the similar.

Legend of the 6 supplemental tables :

- = absent, + = present, NA = Not Available, ARVC = Arrhythmogenic Right Ventricular Cardiomyopathy, *CDSN* = Corneodesmosin gene, *DSC2* = Desmocollin 2 gene, *DSC3* = Desmocollin 3 gene, *DSG1* = Desmoglein 1 gene, *DSG4* = Desmoglein 4 gene, *DSP* = Desmoplakin gene, *JUP* = Plakoglobin gene, *PKP1* = Plakophillin 1 gene, PPK = Palmoplantar Keratoderma, PTC = Premature Termination Codon

References :

1. McKoy, G. *et al.* Identification of a deletion in plakoglobin in arrhythmogenic right ventricular cardiomyopathy with palmoplantar keratoderma and woolly hair (Naxos disease). *Lancet* 355, 2119–2124 (2000).
2. Protonotarios, N. *et al.* Genotype-phenotype assessment in autosomal recessive arrhythmogenic right ventricular cardiomyopathy (Naxos disease) caused by a deletion in plakoglobin. *J. Am. Coll. Cardiol.* 38, 1477–1484 (2001).
3. Narin, N. *et al.* Arrhythmogenic right ventricular cardiomyopathy (Naxos disease): report of a Turkish boy. *Pacing Clin. Electrophysiol. PACE* 26, 2326–2329 (2003).
4. Antoniades, L. *et al.* Arrhythmogenic right ventricular cardiomyopathy caused by deletions in plakophilin-2 and plakoglobin (Naxos disease) in families from Greece and Cyprus: genotype-phenotype relations, diagnostic features and prognosis. *Eur. Heart J.* 27, 2208–2216 (2006).
5. Lazaros, G. *et al.* Naxos disease presenting with ventricular tachycardia and troponin elevation. *Heart Vessels* 24, 63–65 (2009).
6. Erken, H. *et al.* Cardiomyopathy with alopecia and palmoplantar keratoderma (CAPK) is caused by a JUP mutation. *Br. J. Dermatol.* 165, 917–921 (2011).
7. Cabral, R. M. *et al.* Homozygous mutations in the 5' region of the JUP gene result in cutaneous disease but normal heart development in children. *J. Invest. Dermatol.* 130, 1543–1550 (2010).
8. Pigors, M. *et al.* Lack of plakoglobin leads to lethal congenital epidermolysis bullosa: a novel clinico-genetic entity. *Hum. Mol. Genet.* 20, 1811–1819 (2011).
9. Whittock, N. V. *et al.* Striate palmoplantar keratoderma resulting from desmoplakin haploinsufficiency. *J. Invest. Dermatol.* 113, 940–946 (1999).

10. Armstrong, D. K. *et al.* Haploinsufficiency of desmoplakin causes a striate subtype of palmoplantar keratoderma. *Hum. Mol. Genet.* 8, 143–148 (1999).
11. Boulé, S. *et al.* Expanding the phenotype associated with a desmoplakin dominant mutation: Carvajal/Naxos syndrome associated with leukonychia and oligodontia. *Int. J. Cardiol.* 161, 50–52 (2012).
12. Keller, D. I. *et al.* De novo heterozygous desmoplakin mutations leading to Naxos-Carvajal disease. *Swiss Med. Wkly.* 142, w13670 (2012).
13. Norgett, E. E. *et al.* Early death from cardiomyopathy in a family with autosomal dominant striate palmoplantar keratoderma and woolly hair associated with a novel insertion mutation in desmoplakin. *J. Invest. Dermatol.* 126, 1651–1654 (2006).
14. Chalabreysse, L. *et al.* A new hypo/oligodontia syndrome: Carvajal/Naxos syndrome secondary to desmoplakin-dominant mutations. *J. Dent. Res.* 90, 58–64 (2011).
15. Norgett, E. E. *et al.* Recessive mutation in desmoplakin disrupts desmoplakin-intermediate filament interactions and causes dilated cardiomyopathy, woolly hair and keratoderma. *Hum. Mol. Genet.* 9, 2761–2766 (2000).
16. Uzumcu, A. *et al.* Loss of desmoplakin isoform I causes early onset cardiomyopathy and heart failure in a Naxos-like syndrome. *J. Med. Genet.* 43, e5 (2006).
17. Christensen, A. H. *et al.* Wide spectrum of desmosomal mutations in Danish patients with arrhythmogenic right ventricular cardiomyopathy. *J. Med. Genet.* 47, 736–744 (2010).
18. Krishnamurthy, S. *et al.* Arrhythmogenic dilated cardiomyopathy due to a novel mutation in the desmoplakin gene. *Indian J. Pediatr.* 78, 866–869 (2011).

19. Williams, T. *et al.* Novel desmoplakin mutation: juvenile biventricular cardiomyopathy with left ventricular non-compaction and acantholytic palmoplantar keratoderma. *Clin. Res. Cardiol. Off. J. Ger. Card. Soc.* 100, 1087–1093 (2011).

20. Rasmussen, T. B. *et al.* Protein expression studies of desmoplakin mutations in cardiomyopathy patients reveal different molecular disease mechanisms. *Clin. Genet.* 84, 20–30 (2013).

21. Yesudian, P. D. *et al.* Novel compound heterozygous mutations in the desmoplakin gene cause hair shaft abnormalities and culminate in lethal cardiomyopathy. *Clin. Exp. Dermatol.* 39, 506–508 (2014).

22. Pigors, M. *et al.* Desmoplakin mutations with palmoplantar keratoderma, woolly hair and cardiomyopathy. *Acta Derm. Venereol.* 95, 337–340 (2015).

23. Alcalai, R., Metzger, S., Rosenheck, S., Meiner, V. & Chajek-Shaul, T. A recessive mutation in desmoplakin causes arrhythmogenic right ventricular dysplasia, skin disorder, and woolly hair. *J. Am. Coll. Cardiol.* 42, 319–327 (2003).

24. Molho-Pessach, V. *et al.* Two Novel Homozygous Desmoplakin Mutations in Carvajal Syndrome. *Pediatr. Dermatol.* (2015). doi:10.1111/pde.12541

25. Asimaki, A. *et al.* Unique epidermolytic bullous dermatosis with associated lethal cardiomyopathy related to novel desmoplakin mutations. *J. Cutan. Pathol.* 36, 553–559 (2009).

26. Vahlquist, A. *et al.* A Scandinavian case of skin fragility, alopecia and cardiomyopathy caused by DSP mutations. *Clin. Exp. Dermatol.* 39, 30–34 (2014).

27. Antonov, N. K. *et al.* Early-onset heart failure, alopecia, and cutaneous abnormalities associated with a novel compound heterozygous mutation in desmoplakin. *Pediatr. Dermatol.* 32, 102–108 (2015).

28. Mahoney, M. G. *et al.* Compound heterozygous desmoplakin mutations result in a phenotype with a combination of myocardial, skin, hair, and enamel abnormalities. *J. Invest. Dermatol.* 130, 968–978 (2010).

29. Tanaka, A. *et al.* Novel truncating mutations in PKP1 and DSP cause similar skin phenotypes in two Brazilian families. *Br. J. Dermatol.* 160, 692–697 (2009).

30. Whittock, N. V. *et al.* Compound heterozygosity for non-sense and mis-sense mutations in desmoplakin underlies skin fragility/woolly hair syndrome. *J. Invest. Dermatol.* 118, 232–238 (2002).

31. Al-Owain, M. *et al.* Novel homozygous mutation in DSP causing skin fragility-woolly hair syndrome: report of a large family and review of the desmoplakin-related phenotypes. *Clin. Genet.* 80, 50–58 (2011).

32. Smith, F. J. D., Wilson, N. J., Moss, C., Dopping-Hepenstal, P. & McGrath, J. Compound heterozygous mutations in desmoplakin cause skin fragility and woolly hair. *Br. J. Dermatol.* 166, 894–896 (2012).

33. Jonkman, M. F. *et al.* Loss of desmoplakin tail causes lethal acantholytic epidermolysis bullosa. *Am. J. Hum. Genet.* 77, 653–660 (2005).

34. Bolling, M. C. *et al.* Lethal acantholytic epidermolysis bullosa due to a novel homozygous deletion in DSP: expanding the phenotype and implications for desmoplakin function in skin and heart. *Br. J. Dermatol.* 162, 1388–1394 (2010).

35. Hobbs, R. P. *et al.* Insights from a desmoplakin mutation identified in lethal acantholytic epidermolysis bullosa. *J. Invest. Dermatol.* 130, 2680–2683 (2010).

36. McGrath, J. A. *et al.* Skin fragility and hypohidrotic ectodermal dysplasia resulting from ablation of plakophilin 1. *Br. J. Dermatol.* 140, 297–307 (1999).

37. Sprecher, E. *et al.* Homozygous splice site mutations in PKP1 result in loss of epidermal plakophilin 1 expression and underlie ectodermal dysplasia/skin fragility syndrome in two consanguineous families. *J. Invest. Dermatol.* 122, 647–651 (2004).

38. Abdalla, E. M. & Has, C. A plakophilin-1 gene mutation in an egyptian family with ectodermal dysplasia-skin fragility syndrome. *Mol. Syndromol.* 5, 304–306 (2014).

39. Ersoy-Evans, S. *et al.* Ectodermal dysplasia-skin fragility syndrome resulting from a new homozygous mutation, 888delC, in the desmosomal protein plakophilin 1. *J. Am. Acad. Dermatol.* 55, 157–161 (2006).

40. Boyce, A. E. *et al.* Ectodermal dysplasia-skin fragility syndrome due to a new homozygous internal deletion mutation in the PKP1 gene. *Australas. J. Dermatol.* 53, 61–65 (2012).

41. McGrath, J. A. *et al.* Mutations in the plakophilin 1 gene result in ectodermal dysplasia/skin fragility syndrome. *Nat. Genet.* 17, 240–244 (1997).

42. Zheng, R., Bu, D.-F. & Zhu, X.-J. Compound heterozygosity for new splice site mutations in the plakophilin 1 gene (PKP1) in a Chinese case of ectodermal dysplasia-skin fragility syndrome. *Acta Derm. Venereol.* 85, 394–399 (2005).

43. Whittock, N. V. *et al.* Genomic amplification of the human plakophilin 1 gene and detection of a new mutation in ectodermal dysplasia/skin fragility syndrome. *J. Invest. Dermatol.* 115, 368–374 (2000).

44. Hernández-Martín, A. *et al.* Ectodermal dysplasia-skin fragility syndrome: a novel mutation in the PKP1 gene. *Clin. Exp. Dermatol.* 38, 787–790 (2013).

45. Steijlen, P. M. *et al.* Cryptic splicing at a non-consensus splice-donor in a patient with a novel mutation in the plakophilin-1 gene. *J. Invest. Dermatol.* 122, 1321–1324 (2004).

46. Hamada, T. *et al.* Genotype-phenotype correlation in skin fragility-ectodermal dysplasia syndrome resulting from mutations in plakophilin 1. *Exp. Dermatol.* 11, 107–114 (2002).

47. Rickman, L. *et al.* N-terminal deletion in a desmosomal cadherin causes the autosomal dominant skin disease striate palmoplantar keratoderma. *Hum. Mol. Genet.* 8, 971–976 (1999).

48. Hunt, D. M. *et al.* Spectrum of dominant mutations in the desmosomal cadherin desmoglein 1, causing the skin disease striate palmoplantar keratoderma. *Eur. J. Hum. Genet. EJHG* 9, 197–203 (2001).

49. Keren, H., Bergman, R., Mizrachi, M., Kashi, Y. & Sprecher, E. Diffuse nonepidermolytic palmoplantar keratoderma caused by a recurrent nonsense mutation in DSG1. *Arch. Dermatol.* 141, 625–628 (2005).

50. Dua-Awereh, M. B., Shimomura, Y., Kraemer, L., Wajid, M. & Christiano, A. M. Mutations in the desmoglein 1 gene in five Pakistani families with striate palmoplantar keratoderma. *J. Dermatol. Sci.* 53, 192–197 (2009).

51. Milingou, M., Wood, P., Masouyé, I., McLean, W. H. & Borradori, L. Focal palmoplantar keratoderma caused by an autosomal dominant inherited mutation in the desmoglein 1 gene. *Dermatol. Basel Switz.* 212, 117–122 (2006).

52. Kljuic, A. *et al.* A nonsense mutation in the desmoglein 1 gene underlies striate keratoderma. *Exp. Dermatol.* 12, 523–527 (2003).

53. Zamiri, M. *et al.* Mutation in DSG1 causing autosomal dominant striate palmoplantar keratoderma. *Br. J. Dermatol.* 161, 692–694 (2009).

54. Hershkovitz, D., Lugassy, J., Indelman, M., Bergman, R. & Sprecher, E. Novel mutations in DSG1 causing striate palmoplantar keratoderma. *Clin. Exp. Dermatol.* 34, 224–228 (2009).

55. Barber, A. G., Wajid, M., Columbo, M., Lubetkin, J. & Christiano, A. M. Striate palmoplantar keratoderma resulting from a frameshift mutation in the desmoglein 1 gene. *J. Dermatol. Sci.* 45, 161–166 (2007).

56. Samuelov, L. *et al.* Desmoglein 1 deficiency results in severe dermatitis, multiple allergies and metabolic wasting. *Nat. Genet.* 45, 1244–1248 (2013).

57. Has, C. *et al.* Loss of desmoglein 1 associated with palmoplantar keratoderma, dermatitis and multiple allergies. *Br. J. Dermatol.* 172, 257–261 (2015).

58. Kljuic, A. *et al.* Desmoglein 4 in hair follicle differentiation and epidermal adhesion: evidence from inherited hypotrichosis and acquired pemphigus vulgaris. *Cell* 113, 249–260 (2003).

59. Rafiq, M. A. *et al.* A recurrent intragenic deletion mutation in DSG4 gene in three Pakistani families with autosomal recessive hypotrichosis. *J. Invest. Dermatol.* 123, 247–248 (2004).

60. John, P. *et al.* Recurrent intragenic deletion mutation in desmoglein 4 gene underlies autosomal recessive hypotrichosis in two Pakistani families of Balochi and Sindhi origins. *Arch. Dermatol. Res.* 298, 135–137 (2006).

61. Messenger, A. G., Bazzi, H., Parslew, R., Shapiro, L. & Christiano, A. M. A missense mutation in the cadherin interaction site of the desmoglein 4 gene underlies localized autosomal recessive hypotrichosis. *J. Invest. Dermatol.* 125, 1077–1079 (2005).

62. Wajid, M. *et al.* Localized autosomal recessive hypotrichosis due to a frameshift mutation in the desmoglein 4 gene exhibits extensive phenotypic variability within a Pakistani family. *J. Invest. Dermatol.* 127, 1779–1782 (2007).

63. Shimomura, Y., Sakamoto, F., Kariya, N., Matsunaga, K. & Ito, M. Mutations in the desmoglein 4 gene are associated with monilethrix-like congenital hypotrichosis. *J. Invest. Dermatol.* 126, 1281–1285 (2006).

64. Zlotogorski, A. *et al.* An autosomal recessive form of monilethrix is caused by mutations in DSG4: clinical overlap with localized autosomal recessive hypotrichosis. *J. Invest. Dermatol.* 126, 1292–1296 (2006).

65. Schaffer, J. V. *et al.* Mutations in the desmoglein 4 gene underlie localized autosomal recessive hypotrichosis with monilethrix hairs and congenital scalp erosions. *J. Invest. Dermatol.* 126, 1286–1291 (2006).

66. Farooq, M., Ito, M., Naito, M. & Shimomura, Y. A case of monilethrix caused by novel compound heterozygous mutations in the desmoglein 4 (DSG4) gene. *Br. J. Dermatol.* 165, 425–431 (2011).

67. Kato, M. *et al.* An Autosomal Recessive Mutation of DSG4 Causes Monilethrix through the ER Stress Response. *J. Invest. Dermatol.* (2015). doi:10.1038/jid.2015.12

68. Simpson, M. A. *et al.* Homozygous mutation of desmocollin-2 in arrhythmogenic right ventricular cardiomyopathy with mild palmoplantar keratoderma and woolly hair. *Cardiology* 113, 28–34 (2009).

69. Ayub, M. *et al.* A homozygous nonsense mutation in the human desmocollin-3 (DSC3) gene underlies hereditary hypotrichosis and recurrent skin vesicles. *Am. J. Hum. Genet.* 85, 515–520 (2009).

70. Levy-Nissenbaum, E. *et al.* Hypotrichosis simplex of the scalp is associated with nonsense mutations in CDSN encoding corneodesmosin. *Nat. Genet.* 34, 151–153 (2003).

71. Dávalos, N. O. *et al.* A non-sense mutation in the corneodesmosin gene in a Mexican family with hypotrichosis simplex of the scalp. *Br. J. Dermatol.* 153, 1216–1219 (2005).

72. Huang, X., Jiang, H. & Quan, Q. [Clinical investigation of a Chinese family with hypotrichosis simplex of the scalp and mutational analysis of CDSN gene]. *Zhonghua Yi Xue Yi Chuan Xue Za Zhi Zhonghua Yixue Yichuanxue Zazhi Chin. J. Med. Genet.* 29, 452–454 (2012).

73. Telem, D. F., Israeli, S., Sarig, O. & Sprecher, E. Inflammatory peeling skin syndrome caused a novel mutation in CDSN. *Arch. Dermatol. Res.* 304, 251–255 (2012).

74. Oji, V. *et al.* Loss of corneodesmosin leads to severe skin barrier defect, pruritus, and atopy: unraveling the peeling skin disease. *Am. J. Hum. Genet.* 87, 274–281 (2010).

75. Mallet, A. *et al.* Identification of the first nonsense CDSN mutation with expression of a truncated protein causing peeling skin syndrome type B. *Br. J. Dermatol.* 169, 1322–1325 (2013).

76. Israeli, S., Zamir, H., Sarig, O., Bergman, R. & Sprecher, E. Inflammatory peeling skin syndrome caused by a mutation in CDSN encoding corneodesmosin. *J. Invest. Dermatol.* 131, 779–781 (2011).

77. Ishida-Yamamoto, A. *et al.* Inflammatory peeling skin syndrome caused by homozygous genomic deletion in the PSORS1 region encompassing the CDSN gene. *Exp. Dermatol.* 23, 60–63 (2014).

78. Teye, K. *et al.* Homozygous deletion of six genes including corneodesmosin on chromosome 6p21.3 is associated with generalized peeling skin disease. *J. Dermatol. Sci.* 75, 36–42 (2014).