

Table S1. Desmoplakin: mutation constellations and skin related phenotypes

No.	Mutation constellation		Location in gene	Inheritance	Original designation, reference	Skin fragility	Woolly hair/alopecia	PPK	ARVC/D	Age of death
	c. DNA change	Amino acid change								
1	c.939+1A>G		Intron 7	Dominant	Striate PPK (17), LDAC (18), ARVC (19)	-	-	+	-	
2	c.991C>T	p.Q331*	Exon 8	Dominant	Striate PPK (20)	-	-	+	-	
3	c.1691C>T	p.T564I	Exon 13	Dominant	Naxos-Carvajal syndrome with leukonychia, oligodontia (21)	-	+	+	-	
4	c.1748 T>C	p.L583P	Exon 14	Dominant	Naxos-Carvajal syndrome with leukonychia, oligodontia (22)	-	+	+	-	
5	c.1790 C>T	p.S597L	Exon 14	Dominant	Naxos-Carvajal syndrome with hypo/oligodontia (23)	-	+	+	-	
6	30 bp insertion	p.I608_	Exon 14	Dominant	Naxos-Carvajal syndrome with absence of premolars (24)	-	+	+	18 years	
7	c.861G>T	p.N287K	Exon 7	Recessive	Skin fragility/woolly hair syndrome (11)	+	+	-	-	
8	c.2427T>A	p.C809*	Exon 17	Recessive	Skin fragility/woolly hair syndrome (11)	+	+	-	-	
9	c.1067C>A	p.T356K	Exon 9	Recessive (?)	Carvajal syndrome (this study)	-	+	+	-	
10	c.2131_2132delAG	p.S711Cfs*4	Exon 16	Recessive	Skin fragility/woolly hair syndrome (11)	+	+	-	-	
11	c.1990C>T	p.Q664*	Exon 15	Recessive	Skin fragility/woolly hair syndrome (11)	+	+	-	-	
12	c.7096C>T	p.R2366C	Exon 24	Recessive	Skin fragility/woolly hair syndrome (11)	+	+	-	-	
13	c.2516_2519delACTC	p.H839fs*23	Exon 18	Recessive	Ectodermal dysplasia/skin fragility syndrome (25)	+	+	-	-	
14	c.3971_3974delATAA	p.N1324fs*23	Exon 23	Recessive	Ectodermal dysplasia/skin fragility syndrome (25)	+	+	-	-	
15	c.2876_2877+3del5	p.L959Mfs*5	Exon/intron20	Recessive	Ectodermal dysplasia/skin fragility syndrome (25)	+	+	-	-	
16	c.3799C>T	p.R1267*	Exon 23	Recessive	Ectodermal dysplasia/skin fragility syndrome (25)	+++	-	+/-	Neonatal	
17	c.3901C>T	p.Q1301*	Exon 23	Recessive	Acantholytic epidermolysis bullosa (26)	-	+	+	3 years	
18	c.4336C>T	p.Q1446*	Exon 23	Recessive	Carvajal syndrome (28)	-	+	+	12, 14 years	
19	c.2017C>T	p.Q673*	Exon 15	Recessive	Epidermolytic bullous dermatitis with ARVC/D (29)	+	+	+	9 years	
20	c.4778_4790del	p.K1593Sfs*5	Exon 23	Recessive	Epidermolytic bullous dermatitis with ARVC/D (29)	+	+	+	-	
21	c.6310delA	p.T2104Qfs*12	Exon 24	Recessive	Skin fragility, alopecia, PPK, ARVC/D (30)	+	+	+	-	
22	c.5208_5209delAG	p.G1737Tfs*7	Exon 24	Recessive	Skin fragility, alopecia, PPK, ARVC/D (30)	-	+	+	6 years	
23	c.5800C>T	p.R1934*	Exon 24	Recessive	Naxos-Carvajal syndrome (31)	+++	-	-	Neonatal	
24	c.6091_6092delTT	p.L2031Gfs*29	Exon 24	Recessive	Acantholytic epidermolysis bullosa (6)	-	+	+	-	
25	c.7567_7570delAAGA	p.E2193K	Exon 24	Recessive	Acantholytic epidermolysis bullosa (6)	-	+	+	-	
26	c.6721_6722delAT	p.K2523Dfs*37	Exon 24	Recessive	Hair shaft abnormalities and in lethal cardiomyopathy (32)	-	+	+	5 years	
27	c.7096C>T	p.R2366C	Exon 24	Recessive	Hair shaft abnormalities and in lethal cardiomyopathy (32)	+	+	-	-	
28	c.7097G>A	p.R2366H	Exon 24	Recessive	Skin fragility/woolly hair syndrome (33)	+	+	-	-	
29	c.7123G>C	p.G2375R	Exon 24	Recessive	Skin fragility/woolly hair syndrome (33)	+	+	-	-	
30	c.7248delIT	p.F2416Lfs*14	Exon 24	Recessive	Skin fragility/woolly hair syndrome (34)	+	+	-	-	
31	c.7566_7567delinsC	p.R2522Sfs*39	Exon 24	Recessive	Skin fragility/woolly hair syndrome (34)	-	+	+	-	
32	c.7756C>T	p.R2586*	Exon 24	Recessive	ARVC/D, woolly hair, PPK (35)	+++	-	-	23 years	
33	c.7623delG	p.K2542Sfs*19	Exon 24	Recessive	Acantholytic epidermolysis bullosa (9)	-	+	+	Neonatal	
34	c.7964C>A	p.A2655D	Exon 24	Recessive	PPK, woolly hair, hypotrichosis (this study)	-	+	-	-	
35	c.6310delA	p.T2104Qfs*12	Exon 24	Recessive	PPK, woolly hair, hypotrichosis (this study)	-	+	-	-	
36	c.7780delIT	p.S2594Pfs*8	Exon 24	Recessive	Naxos-Carvajal syndrome (8)	-	+	+	12-17 years	
37			Exon 24	Recessive	Mucocutaneous blistering, woolly hair, PPK, ARVC/D (36)	+	+	+	14 years	
38			Exon 24	Recessive	Carvajal syndrome (37)	-	+	+	-	

ARVC: arrhythmogenic right ventricular cardiomyopathy; ARVC/D: arrhythmogenic right ventricular dysplasia /cardiomyopathy; LDAC: left-dominant arrhythmogenic cardiomyopathy; PPK: palmoplantar keratoderma; +: present; +++: very severe; +/-: uncertain finding; -: not present.