

Supplementary Tables

Supplementary Table 1. Desmosomal mutations represented in the study population

Gene; n (%)	Amino Acid Change	Nucleotide Change	Overall n=87 (# families)
PKP2; 76 (87)	Mutant splice product	2146-1G->C	18 (9)
	Mutant splice product	2489+1G>A	10 (5)
	A733fsX740	2197_2202delinsG	7 (4)
	W538X	1613G>A	6 (4)
	R79X	235C>T	5 (4)
	P672fsX683	2013delC	5 (1)
	V837fsX930	2509delA	4 (2)
	Mutant splice product	337-2A>T	4 (1)
	P323fsX350	968_971delAGGC	3 (1)
	S587fsX69	1759delG	3 (1)
	Q617X	1849C>T	2 (1)
	S406fsX4	1211-1212insT	1 (1)
	F424S	1271T>C	1 (1)
	Splicing change	2145+1G>A	1 (1)
	Mutant splice product	224-3C>G	1 (1)
	R413X	1237C>T	1 (1)
	S50fsX110	148_151delACAG	1 (1)
	L787F	2359C>T	1 (1)
	Q74fsX85	218dupG	1 (1)
V548fsX562	1643delG	1 (1)	
DSG2; 7 (8)	R46Q	137G>A	2 (1)
	C507Y	1520G>A	2 (1)
	G812C	2434G>T	1 (1)
	K346del	1038_1040delGAA	1 (1)
	T335A	1003A>G	1 (1)
DSP; 3 (3)	E422K	1264G>A	1 (1)
	R2160X	6478C>T	1 (1)
	V30M	88G>A	1 (1)
DSC2; 1 (1)	E426K	1276G>A	1 (1)

Abbreviations: *DSC2*: Desmocollin-2, *DSG2*: desmoglein-2, *DSP*: Desmoplakin, *PKP2*: Plakophilin-2