Table SI. PCR details of a multi-gene panel test.

System	Reagent	Thermocycling conditions (°C, time)	No. of cycles	Source of DNA	Bullous disease-associated genes	Primer sequences
DNA library construction PCR system	25 μl 2x HiFi HotStart ReadyMix 5 μl Pre-LM-PCR Oligos 1&2.5 μM	98, 45 sec 98, 45 sec 60, 30 sec 72, 30 sec 72, 1 min 10, end	9	Adaptor-Ligated DNA	KRT5, KRT14, PLEC, DST, KLHL24, TGM5, DSP, PKP1, JUP, EXPH5, COL7A1, LAMA3, LAMB3, LAMC2, ITGA6, ITGB4, COL17A1, CD151, ITGA3, PLCG2 and FERMT1	Forward 5'-AATGATACGGCGACCACCGA; Reverse 5'-CAAGCAGAAGACGGCATACGA
Target area capture PCR system	25 μl 2x HiFi HotStart ReadyMix 5 μl Post-LM-PCR Oligos 1&2.5 μM	98, 45 sec 98, 15 sec 60, 30 sec 72, 30 sec 72, 1 min 10, end	10	Captured DNA Library	KRT5, KRT14, PLEC, DST, KLHL24, TGM5, DSP, PKP1, JUP, EXPH5, COL7A1, LAMA3, LAMB3, LAMC2, ITGA6, ITGB4, COL17A1, CD151, ITGA3, PLCG2 and FERMT1	Forward 5'-AATGATACGGCGACCACCGA; Reverse 5'-CAAGCAGAAGACGGCATACGA

Table SII. Summary of fermitin family member 1mutations in Kindler syndrome.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
1	Has, 2015, Clinical Genetics	Noncoding exon 1	c.-20A>G	Reduced transcription	-	1	Italian	(42)
2	Jobard, 2003 Human Molecular Genetics; Has, 2015, Clinical Genetics	Promoter region and noncoding exon 1	g.-711-1241del	Reduced transcription	Deletion	2	Italian; Druze	(8,42)
3	Has, 2015, Clinical genetics	Promoter region and the 5' region	g.6140393_6102171 delinsCAAACTGA	-	Deletion	1	Indian	(42)
4	Sadler, 2006, Archives of Dermatology	Exon 2	20/21delTT	p.Phe7fsX11	Frameshift	1	Turkish	(43)
5	Kantheti, 2017, Clinical and Experimental Dermatology	Exon 2	c.21delT	p.Phe7fs24X	Frameshift	1	Indian	(44)
6	Techanukul, 2011, Acta Dermato-Venereologica	Exon 2	-	p.Trp12X	Nonsense	1	Indian	(22)
7	Has, 2006,The Journal of Investigative Dermatology	Exon 2	c.95_96delGA	p.Arg32fsX63	Frameshift	1	Italian	(32)
8	Fuchs-Telem, 2014, Clinical and Experimental Dermatology; El Hachem, 2015, European Journal of Dermatology	Exon 2	c.137_140delTAGT	p.Leu46X	Frameshift	2	Arab Muslim; Palestinian	(8,45)
9	Techanukul, 2015, Acta Dermato-Venereologica	Exon 2	-	p.Gln49X	Nonsense	1	Australian	(22)
10	Has, 2011, Human Mutation	Exon 3	c.152-2delAGinsCT	AS	Frameshift	1	-	(46)
11	Mansur, 2007, Acta Dermato-Venereologica	Exon 3	c.170C >A	p.Ser57X	Nonsense	1	Turkish	(7)
12	Niculescu, 2018, Hautarzt; Lennartz, 2012, Journal of the German Society of Dermatology	Exon 3	c.190G> T	p.Glu64X	Nonsense	2	German; Turkish	(47,48)
13	Siegel, 2003, American journal of Human Genetics	Exon 3	c.193C>T	p.Gln65X	Nonsense	1	Jordanian	(38)
14	Has, 2011, Human Mutation	Exon 3	c.299_301del	p.Arg100del	Frameshift	1	-	(46)
15	Lai-Cheong, 2009, The British Journal of Dermatology; Youssefian, 2015, The Journal of Investigative Dermatology; Has, 2009, The American Journal of Pathology; Almeida, 2013, Anais Brasileiros de Dermatologia; Has, 2010, Dermatology	Exon 3	c.328C>T	p.Arg110X	Nonsense	5	Iranian; -; Hispanic American; Brazilian; -	(9,20, 23,34,35)

Table SII. Continued.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
16	Lai-Cheong, 2007, The Journal of Investigative Dermatology	Exon 3	c.341C>G	p.Ser114X	Nonsense	1	Indian	(49)
17	Has, 2006, The Journal of Investigative Dermatology; Siegel, 2003, American Journal of Human Genetics	Exon 3	c.373delT	p.Ile124fs	Frameshift	2	Italian; Italian	(32,38)
18	Jobard, 2003, Human Molecular Genetics	Exon 3	c.385+2T>C	-	Splice	1	Algerian	(6)
19	Techanukul, 2011, Acta Dermato-Venereologica	Exon 3	c.384_385+2del4	-	Splice	1	Indian	(22)
20	Valinotto, 2020, Pediatric Dermatology	Exon 4	c.450delG	p. Asp153ThrfsX8	Frameshift	1	Paraguayan	(50)
21	Has, 2009, The American Journal of Pathology; Kiritsi, 2012, The Journal of Clinical Investigation	Exon 4	c.456dupA	p.Asp153ArgfsX 4	Frameshift	2	-; German	(23,24)
22	Jobard, 2003, Human Molecular Genetics	Exon 4	c.464delA	p.Asn155fs	Frameshift	1	Algerian	(6)
23	Lai-Cheong, 2007, The Journal of Investigative Dermatology	Exon 4	c.502G>T	p.Glu168X	Nonsense	1	Indian	(49)
24	Youssefian, 2015, The Journal of Investigative Dermatology;	Exon 5	c.550_551insA	p.Ser184LeufsX1	Frameshift	1	Iranian	(20)
25	Youssefian, 2015, The Journal of Investigative Dermatology; Siegel, 2003, American Journal of Human Genetics	Exon 5	c.614G>A	p.Trp205X	Nonsense	2	Iranian; Pakistani	(20,38)
26	Heidari, 2016, Archives of Iranian Medicine	Exon 5	c.676C>T	p.Gln226X	Nonsense	1	Iranian	(51)
27	Mansur, 2007, Acta Dermato-Venereologica; Techanukul, 2011, Acta Dermato-Venereologica; Has, 2009, The American journal of pathology; Kiritsi, 2012, The Journal of Clinical Investigation; Kern, 2007, The Journal of Pathology; Lai-Cheong, 2012, The Journal of Investigative Dermatology; Ashton, 2006, The Journal of Investigative Dermatology; Martignago, 2007, The British Journal of Dermatology; Shaiq, 2012, The Journal of Dermatology; Thomson, 2006, Clinical and Experimental Dermatology; Gkaitatzi, 2009, Oxford Medical Case Reports	Exon 5	c.676dupC/c.676insC	p.Gln226fsX16	Frameshift	11	Serbian, Australian, Indian; Albanian; -; Kosovo Albanian, Turkey, Serbian-Greek; Kosovo Albanian; -; Pakistani; Brazilian; Pakistani; Pakistani; Greek Caucasian	(7,22-31)
28	Chmel, 2015, The Journal of Investigative Dermatology	Exon 5	c.676delC	p.Gln226SerfsX26	Frameshift	1	German	(52)
29	Youssefian, 2015, The Journal of Investigative Dermatology	Exon 5	g.6109607_6112272del	Reduced transcription	Deletion	1	Iranian	(20)
30	Techanukul, 2011, Acta Dermato-Venereologica	Exon 6	c.750G>A	p.Trp250X	Nonsense	1	Jewish-Kurdish	(22)
31	Jobard, 2003, Human Molecular Genetics	Exon 6	c.787C>T	p.Gln263X	Nonsense	1	Senegalese	(6)

Table SII. Continued.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
32	Siegel, 2003, American Journal of Human Genetics; Kantheti, 2017, Clinical and Experimental Dermatology; Burch, 2006, Archives of Dermatology;	Exon 6	c.811C>T	p.Arg271X	Nonsense	3	Panamanian, American, Omanese; Indian; Caucasian	(38,44,53)
33	Siegel, 2003, American Journal of Human Genetics; Krishna, 2014, Clinical and Experimental Dermatology; Atzori, 2018, Journal of the European Academy of Dermatology and Venereology	Exon 7	c.862C>T	p.Arg288X	Nonsense	3	British, Turkish; Indian; Caucasian	(38,54,55)
34	Almeida, 2013, Anais Brasileiros de Dermatologia;	Exon 7	c.866insGG	p.Ala289GlufsX7	Frameshift	1	Brazilian	(34)
35	Youssefian, 2015, The Journal of Investigative Dermatology	Exon 7	c.889A>G	p.Arg297Gly	Missense	1	Iranian	(20)
36	Ashton, 2006, The Journal of Investigative Dermatology	Exon 7	c.905T>A	p.Leu302X	Nonsense	1	British Caucasian	(27)
37	Youssefian, 2015, The Journal of Investigative Dermatology; Has, 2009, The American journal of pathology; Kern, 2007, The Journal of Pathology; Ashton, 2006, The Journal of Investigative Dermatology; Has, 2006, The Journal of Investigative Dermatology; Has, 2004, The Journal of Investigative Dermatology	Exon 7	c.910G>T	p.Glu304X	Nonsense	6	Iranian; -; Australian; British Caucasian; Italian; Caucasian	(20,23,25,27,32,33)
38	Youssefian, 2015, The Journal of Investigative Dermatology	Intron 7	c.957+1G>A	AS	Splice	1	Iranian	(20)
39	Ashton, 2006, The Journal of Investigative Dermatology	Intron 7	c.958-1G>A/IVS7-1G>A	AS	Splice	1	Italian	(27)
40	Oh, 2016, Annals of Dermatology; Youssefian, 2015, The Journal of Investigative Dermatology	Exon 8	c.994_995delCA	p.Gln332GlyfsX9	Frameshift	2	Chinese; Iranian	(17,20)
41	Has, 2011, Human Mutation	Exon 8	c.1051G>T	p.Glu351X	Nonsense	1	-	(46)

Table SII. Continued.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
42	Siegel, 2003, American Journal of Human Genetics; Wada, 2012, The Journal of Dermatology; Natsuga, 2011, Journal of Dermatological Science	Exon 8	c.1089delG/ IVS8+1delG	p.Leu363fs/IVS8+1delG	Frameshift or splice	3	Japanese; Japanese; Japanese	(38-40)
43	Chmel, 2015, The Journal of Investigative Dermatology	Intron 9	c.1139+740G>A/IVS9+740G>A	p.Pro381HisfsX16	Splice	1	German, Romanian, Afghanistan	(52)
44	Youssefian, 2015, The Journal of Investigative Dermatology	Intron 9	c.1139+2T>C	AS	Splice	1	Iranian	(20)
45	Arita, 2007, The British Journal of Dermatology	Intron 9	c.1140-6T>A/IVS9-6T>A	AS	Splice	1	Indian	(56)
46	Kartal, 2016, Journal of the European Academy of Dermatology and Venereology	Intron 9	c.1140-2A>T	AS	Splice	1	Turkish	(12)
47	Ashton, 2006, The Journal of Investigative Dermatology; Has, 2006, The Journal of Investigative Dermatology	Exon 10	c.1161delA	p.Ala388LeufsX14	Frameshift	2	British Caucasian; Italian	(27,32)
48	Youssefian, 2015, The Journal of Investigative Dermatology	Exon 10	c.1176T>G	p.Tyr392X	Nonsense	1	Iranian	(20)
49	Roda, 2018, Dermatology Online Journal	Exon 10	c.1179G>A	p.Trp393X	Nonsense	1	-	(13)
50	Has, 2011, Human Mutation	Exon 10	c.1198T>C	p.Ser400Pro	Missense	1	-	(46)
51	Techanukul, 2011, Acta Dermato-Venereologica; Has, 2009, The American Journal of Pathology	Exon 10	c.1209C>G	p.Tyr403X	Nonsense	2	Arabic; -	(22,23)
52	Lai-Cheong, 2009, The British Journal of Dermatology; Has, 2009, The American Journal of Pathology	Exon 10	c.1217dupA/c.1217insA	p.Asn406LysfsX1	Frameshift	2	-; Canadian	(9,23)
53	Has, 2011, Human Mutation	Exon 10	c.1264+1G>A	AS	Splice	1	-	(46)
54	Zheng, 2019, JAAD case reports	Exon 11	c.1343T>A	p.Met448Lys	Missense	1	Chinese	(19)
55	Has, 2009, The American Journal of Pathology; Mas-Vidal, 2010, Journal of the European Academy of Dermatology and Venereology	Exon 11	c.1365_1371+3del10	Truncated kindlin-1 protein	Splice	2	-; Spanish	(23,57)

Table SII. Continued.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
56	Has, 2011, Human Mutation	Exon 11	c.1371+4A>G	AS	Splice	1	-	(46)
57	Has, 2011, Human mutation	Exon 12	c.1378C>T	p.Gln460X	Nonsense	1	-	(46)
58	Youssefian, 2015, The Journal of Investigative Dermatology	Exon 12	c.1383C>A	p.Tyr461X	Nonsense	1	Iranian	(20)
59	Sethuraman, 2005, Clinical and Experimental Dermatology	Exon 12	c.1404C>A	p.Cys468X	Nonsense	1	Indian	(58)
60	Lai-Cheong, 2007, The Journal of Investigative Dermatology	Exon 12	c.1546G>T	p.Glu516X	Nonsense	1	Indian	(49)
61	Shimizu, 2014, Acta Dermato-Venereologica	Exon 12	c.1564delC	-	Frameshift	1	-	(59)
62	Has, 2011, Human Mutation	Exon 13	c.1675T>C	p.Trp559Arg	Missense	1	-	(46)
63	Kiritsi, 2012, The Journal of Clinical Investigation	Exon 13	c.1677G>A	p.Trp559X	Nonsense	1	Serbian-Greek	(24)
64	Jobard, 2003, Human molecular genetics	Exon 13	c.1714insA	p.Val572fs	Frameshift	1	Tunisian	(6)
65	Has, 2009, The American Journal of Pathology; Lai-Cheong, 2007, The Journal of Investigative Dermatology; Has, 2008, The British Journal of Dermatology	Intron 13	c.1718+1G>A/IVS13+1G>A	C-terminal truncation	Splice	3	-; Indian; Somalian	(23,49,60)
66	Youssefian, 2015, The Journal of Investigative Dermatology; Techanukul, 2011, Acta Dermato-Venereologica; Fassihi, 2005, Journal of Dermatological Science; Diociaiuti, 2016, Journal of the European Academy of Dermatology and Venereology	Intron 13	c.1718+2T>C/IVS13+2T>C	AS	Splice	4	Iranian; Indian; Pakistani; Pakistani	(20,22,36,37)
67	Has, 2006, The Journal of Investigative Dermatology;	Intron 13	IVS13-1G>A	AS	Splice	1	Italian	(32)
68	Has, 2011, Human Mutation	Exon 14	c.1729del	p.Ser577AlafsX14	Frameshift	1	-	(46)
69	Kantheti, 2017, Clinical and Experimental Dermatology	Exon 14	c.1731delC	p.Ser577ArgfsX13	Frameshift	1	Indian	(44)
70	Burch, 2006, Archives of Dermatology	Exon 14	c.1755delT	-	Frameshift	1	Caucasian	(53)
71	Natsuga, 2011, Journal of Dermatological Science; Ohashi, 2015, International Journal of Dermatology	Exon 14	c.1761T>A	p.Tyr587X	Nonsense	2	Japanese; Japanese	(40,61)
72	Ashton, 2006, The Journal of Investigative Dermatology	Exon 14	c.1848G>A	p.Trp616X	Nonsense	1	Omani	(27)

Table SII. Continued.

No	Author, year, journal	Location	Genomic variants	Mutation consequence	Type	Fre.	Population	(Refs.)
73	Lai-Cheong, 2009, The British Journal of Dermatology; Kacar, 2008, The British Journal of Dermatology; Yildirim, 2017, The Turkish Journal of Pediatrics	Exon 14	c.1848_1851dupGGAA/c.1851insGGAA	p.Thr618GlyfsX10	Frameshift	3	Turkish; Turkish; Turkish	(9,10,41)
74	Lai-Cheong, 2007, The Journal of Investigative Dermatology	Intron 14	IVS14-1G>C	AS	Splice	1	Somalian	(49)
75	Techanukul, 2011, Acta Dermato-Venereologica	Intron 14	IVS14+2T>C	AS	Splice	1	Indian	(22)
76	Has, 2011, Human Mutation	Exon 15	c.1867_1869del	p.Ile623del	Frameshift	1	-	(46)
77	Meng, 2020, Spandidos	Exon 15	c.1885_1901del	p.Val629fs	Frameshift	1	Chinese	The current study
78	Ashton, 2006, The Journal of Investigative Dermatology	Exon 15	c.1909delA	-	Frameshift	1	British Caucasian	(27)
79	Gao, 2015, Science	Exon 1-7	17252-bp deletion	Truncated kindlin-1 protein	Deletion	1	Chinese	(18)
80	Zhou, 2009, The British Journal of Dermatology	Exon 7-9	g.63601_66617del	Truncated kindlin-1 protein	Deletion	1	Chinese	(14)
81	Has, 2009, The American Journal of Pathology; Has, 2006, The Journal of Investigative Dermatology; Has, 2015, Clinical Genetics	Introns 9-11	g.70250_74168del/IVS9_IVS11del	p.Pro381ArgfsX36	Deletion	3	-; Italian; Italian	(23,32,42)
82	Has, 2008, Journal of Dermatological Science; Saleva, 2018, Journal of the German Society of Dermatology	Exons 13-15	g.80929_89169del	Truncated kindlin-1 protein	Deletion	2	Bulgarian; Bulgarian	(11,62)
83	Fukushi, 2019, International Journal of Dermatology	Exons 10-11	-	Truncated kindlin-1 protein	Deletion	1	Indian	(15)

Fre., frequency; AS, altered splicing; -, no data.

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