Supplemental Table 1. Skin biopsy sites and molecular findings

Patient and designation of the biopsy	Phenotype and biopsy site	Kindlin-1 expression in immunofluorescence staining	Sequence analysis of DNA from microdissected keratinocytes
Patient 1 - P1-1	affected – left upper arm	-	c.456dupA homozygous
Patient 1 - P1-2	unaffected – right upper arm	+	c.456dupA and normal sequence*
Patient 1 - P1-3	unaffected – left upper arm	+	c.456dupA heterozygous and homozygous normal sequence
Patient 1 - P1-4	unaffected – left upper leg	+	c.456dupA heterozygous
Patient 2 - P2-1	affected – right lower leg	-	c.676dupC homozygous
Patient 2 - P2-2	unaffected – right hand	+	c.676dupC heterozygous and homozygous normal sequence
Patient 2 - P2-3	unaffected – right lower leg	+	c.676dupC and normal sequence*

Legend: -, negative staining; +, positive staining; * both mutant and normal sequences were identified by direct sequencing of cloned PCR products. The ratio between mutant and normal sequences was about 1:1.

Supplemental Table 2. SNPs in *FERMT1* and neighbouring regions which were analyzed in DNA from patients' lymphocytes

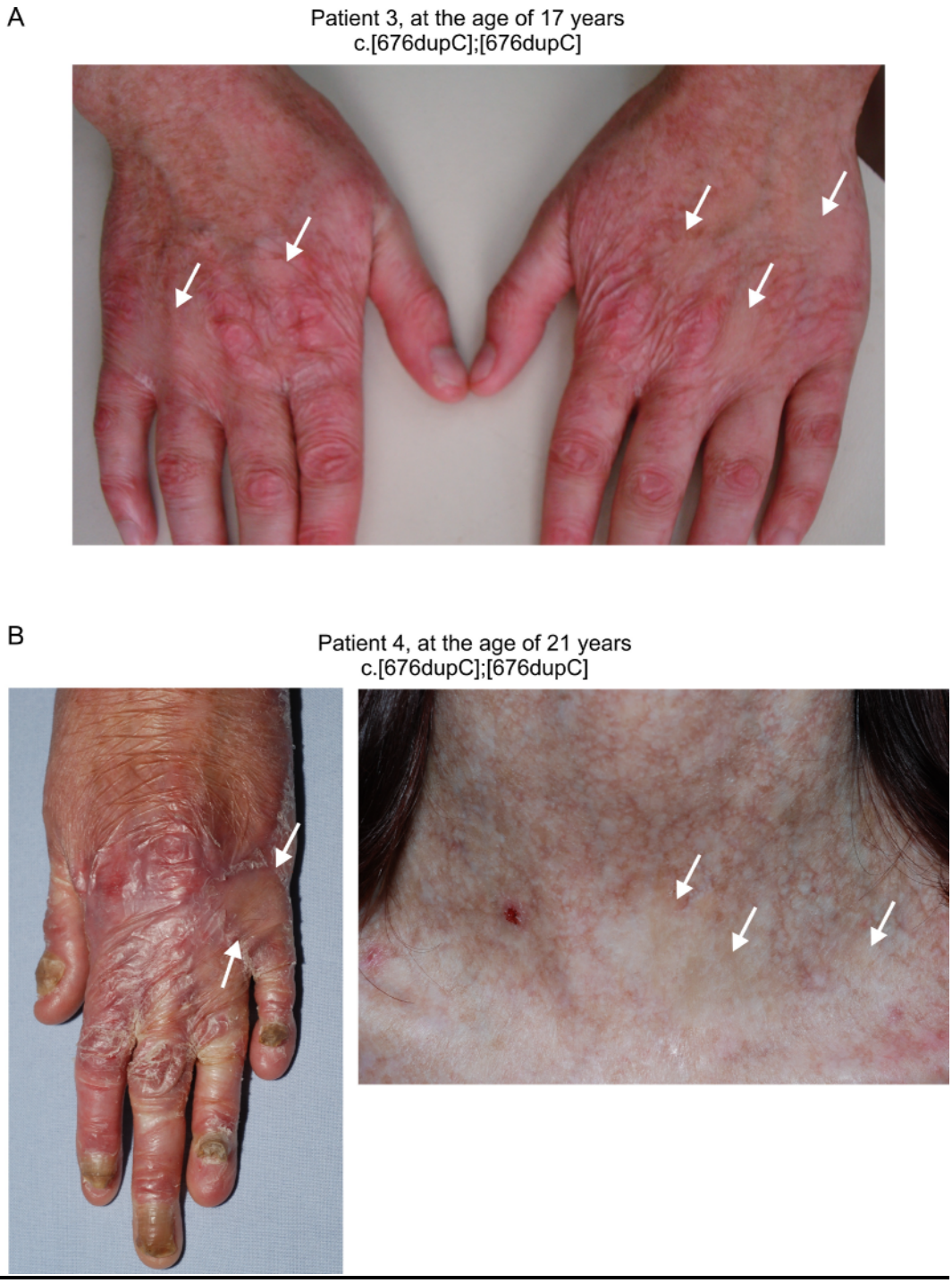
No.	SNP Reference*	Location on chromosome 20	Primer sequence 5'-3'	Sequence variants	
				Patient 1	Patient 2
1.	rs6083461	2,476,778	F: TGACTTTGTGATCCGTCTGC R: CGCTGTCGTTGACTGCTCT	A / T	A / A
2.	rs1178015	2,996,423	F: CCCCATGCTCTCACACTCTT R: CTTTCAGGTGGTAAAAAGTACAGAA	T / T	A / T
3.	rs979332	3,381,044	F: GAAGCCAACAAACACAGCAG R: TCTTGGACACTGCTCACAGC	G / G	A / G
4.	rs2253977	3,939,140	F: GAGAATCGCTTGAACCCAGA R: AAATCTTCCACAATTGGCTCA	A / G	A / G
5.	rs297765	4,440,642	F: TGCACCAGTGTGAATAAGATAGA R: TTTCAAGGTTCGTCCATTCA	A / G	G / G
6.	rs261360	4,991,577	F: TGGAGGTTAGGGTTTCCACA R: GGCTTTTTCACATATCCCTGTT	A / G	A / G
7.	rs881118	5,903,388	F: AGCCTCTATCCCTCCGACAG R: CTCAGAGGTGGCATCTTCCT	A / A	A / A
8.	rs236152	5,903,848	F: GAGGAAGATGCCACCTCTGA R: GCCCCTCTCTTCCTCACTTT	C / C	C / G
9.	rs236153	5,903,894	F: CGACAGGTCCTCTCAAGGAG R: TGTCCTTCACCCAAGAACCT	A / A	A / G
10.	rs6117011	5,922,830	F: CGGCAGCCTCTAAATGTCTT R: CACCACCCCCATAATCTCAG	C / C	C / C
11.	rs454422	5,943,693	F: ACAAGTGCCTGTGTGGCATA R: CAGGCACAGGACACTGAATG	C / C	A / C
12.	rs3897510	6,086,440	F: TCCTTCACTCTCTGCCACCT R: AACCACACCCTCAACCAGAG	T / T	T / T
13.	rs1774886	6,085,395	F: GCAACAAAACCAAGATGTGC R: GCAGGAGGCCTTCCTTATTC	A / A	A / A
14.	rs6038397	6,195,009	F: TGGCTTCTCCATGAATTTCC R: CACCACGTTCTCCCTCAAGT	G / G	G / G
15.	rs10485704	6,188,751	F: AAGGAATTGTAAAACAAGGAGTGAA R: ACACCCAGGTGTCAGGAGAG	T / T	T / T
16.	rs117807751	6,028,870	F: ACTCTTCCACCCCTTTCACC R: GCCAGAGGTATAAATTTGGGAGT	G / G	G / G
17.	rs6085364	6,029,199	F: AGTTCTGGGCCCTCATCTCT R: TCCCCTTCCCTAAAGCCTAA	C / C	C / C
18.	rs78109936	6,029,216	F: CAGATGTGGAGCCAAACTGC R: TTGCAGCAATCCAGATAGGG	T / T	T / T
19.	rs6076920	6,029,317	F: CTCTCCTCCTCCAAGCTTCA R: CTTCCTGGCTAGGCCTTCTT	C / C	C / C
20.	**rs55666319**	**6,090,969**	**F: CACAGTGCCCAGCTTGACT R: CACAATCCCTAGGCCTACCA**	**T / T**	**T / T**
21.	**rs41308641**	**6,093,116**	**F: GGAGGTCTCTGTTCCCCTTT R: GCCTTTCCTCATACAATCAG**	**T / T**	**T / T**
22.	**rs16991866**	**6,093,177**	**F: GGAGGTCTCTGTTCCCCTTT R: GCCTTTCCTCATACAATCAG**	**T / T**	**T / T**
23.	**rs2295435**	**6,096,695**	**F: CTCCAGGACCCTCCAAGCT R: CTTGAAGTAGGCAGAATGCAC**	**G / G**	**G / G**
24.	**rs10373**	**6,100,088**	**F: GGAACCTTGCAGAGTGTTTTG R: GCTCTCCAGGGCATTACAAG**	**T / T**	**T / T**
25.	**rs147571188**	**6,100,252**	**F: GGAACCTTGCAGAGTGTTTTG R: GCTCTCCAGGGCATTACAAG**	**C / C**	**C / C**
26.	rs16991945	6,136,965	F: GACCTGGCTCTCAAGGAAAA R: TACAGTTCTGTGCCCTGTGC	T / T	T / T
27.	rs11907023	6,137,478	F: CCTTCAAAGCATACCACAAGC R: TTTGTGTGGAATTGGTGGTG	C / C	C / C
28.	rs112184907	6,137,505	F: CATCAGCTTCAGTTCACATTACC R: TTTGTGTGGAATTGGTGGTG	ins AA homo	ins AA homo
29.	rs112281423	6,138,928	F: CATGGCTGGATTAATGCTGAG R: GCGCTGTACAGAGGACATGA	G / G	G / G
30.	rs1157366	7,019,980	F: TGTGCACACCATCACTGGTA R: TGTTGAATAAGAAATACGACAGCAA	A / A	A / A
31.	rs2326931	7,398,383	F: TTTCTTCCAATCCATGCACA R: CCCACTTTCACCCCTGTTATT	A / G	A / A
32.	rs6086473	8,484,107	F: AAATGCCAATCTGGTCAAGTG R: CTCAGACCCCCAACCCTATT	C / C	C / T

33.	rs6056209	8,836,771	F: AACCCTTTCTTCCCTTCTGTTC R: CAAGTGATTCACCAGCCTCA	G / G	G / G
34.	rs2072952	9,525,601	F: CCCAGGGTGCAGTATGTTCT R: TGTCTCAACTCCTGCTGCTT	A / G	A / A

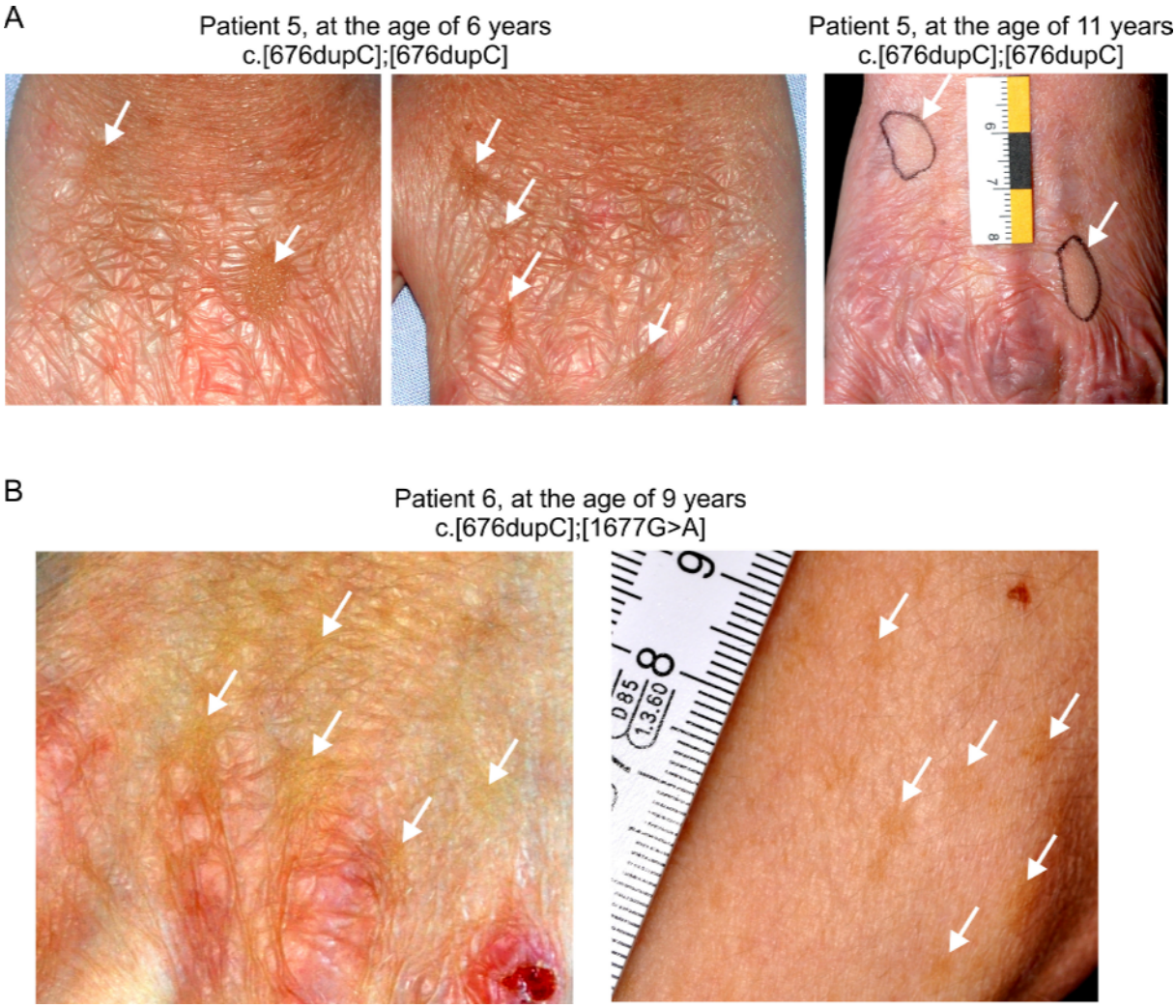
Legend:*, SNP reference ID according to NCBI, framed; the SNP analyzed in microdissected keratinocytes; in bold, SNPs within *FERMT1;* underlined, heterozygous SNPs; grey background, the homozygous interval.

Supplemental Table 3. *FERMT1* primers used in this study for nested PCR

Primer	Sequence 5'-3'
4F	GGAGGTCTCTGTTCCCCTTT
4R	GCCTTTCCTCATCACATCAG
5F	CACAGTGCCCAGCTTGACT
5R	CACAATCCCTAGGCCTACCA
4F nested	GGTCTCTGTTCCCCTTTCCCCTTC
4R nested	GGTGGGGGTGGGAGGAGAGATAT
5F nested	TGGTGCCAAATTTAAAGTCAA
5R nested	ACTGTGTCGGCACTAGCTCA



Supplemental Figure 1. Clinical evidence for revertant mosaicism in patients 3 and 4. Hands of patient 3 at the age of 17 years **(A)** and the left hand and the neck of patient 4 at the age of 21 years **(B)** exhibit normal-appearing skin patches. In each panel, the age at examination and the inherited *FERMT1* mutations are indicated. Areas with clinical features of reversion, namely normal skin texture and absence of atrophy, are pointed out by white arrows. Further clinical features are listed in Table 1.



Supplemental Figure 2. Clinical evidence for revertant mosaicism in patients 5 and 6, two children with KS. **(A)** Patient 5 has been observed since early infancy. The revertant patches were first recognizable at the age of 3 years at the same sites. The hands of patient 5 at the age of 6 years (left and middle panels), and the right hand at the age of 11 years (right panel) are shown. Revertant mosaicism was supported by the stable presence of the marked areas on the right hand. They seem to grow with the child's hand. **(B)** Right hand and left elbow of the 9-year-old patient 6 exhibit multiple normal-appearing, slightly hyperpigmented, skin patches with restored skin texture and absence of atrophy. Further clinical features are listed in Table 1. In each picture, the age at examination and the inherited *FERMT1* mutations are indicated. Areas with clinical features of reversion, namely normal skin texture and absence of skin atrophy, are pointed out by white arrows.