Supp. Table S1. TGM1 Germline Mutations Associated with ARCI

Number	Nucleotide change[a] NM_000359.2	Amino acid change[b] NP_000350	Exon/ intron	Mutation type	References	Alleles[c]
1	c.1-1G>C	ND	Intron 1	Splice Site	This report	Compound heterozygous (c.1-1G>C/c.877-2A>G)
2	c.125C>A	p.Ser42Tyr	Exon 2	Missense	Huber et al. [1995a]	Compound heterozygous (S42Y/R142C/R323Q)
3	c.157T>A	p.Cys53Ser	Exon 2	Missense	Hennies et al. [1998a]	Compound heterozygous (C53S/c.877-2A>G)
4	c.160C>T	p.Arg54X	Exon 2	Nonsense	This report	Homozygous Patient 13
5	c.232C>T	p.Arg78X	Exon 2	Nonsense	Parmentier et al. [1995]	Homozygous
6	c.281G>A	p.Gly94Asp	Exon 2	Missense	Cserhalmi-Friedman et al. [2001]	Heterozygous
7	c.305A>T	p.Asp102Val[d]	Exon 2	Missense	Yang et al. [2001]	Compound heterozygous (D102V/ R307W)
8	c.316C>T	p.Arg106X	Exon 2	Nonsense	Esposito et al. [2007]	Homozygous
9	c.342dupT[e]	Val115CysfsX21	Exon 3	Frameshift	Farasat et al. [2008]	Heterozygous
10	c.370C>T	p.Gln124X	Exon 3	Nonsense	This report	Compound heterozygous (Q124X/R126C)
11	c.374delA[f]	p.Asn125ThrfsX15	Exon 3	Frameshift	Akiyama et al. [2003]	Homozygous
12	c.376C>T	p.Arg126Cys	Exon 3	Missense	Oji et al. [2006]	Compound heterozygous (R126C/R142H)
13	c.377G>A	p.Arg126His	Exon 3	Missense	Farasat et al. [2008]	Compound heterozygous (R126H/L235X)
14	c.379C>T	p.Arg127X	Exon 3	Nonsense	Schorderet et al. [1997]	Homozygous
15	c.401A>G	p.Tyr134Cys	Exon 3	Missense	Farasat et al. [2008]	Compound heterozygous (Y134C/W263X) (Y134C/R396H) (Y134C/c.1331dupA)
16	c.408_411dupCGAC[g]	p.Glu138ArgfsX21	Exon 3	Frameshift	Bichakjian et al. [1998]	Compound heterozygous (c.408_411dupCGAC/ c.1225_1226del AC)
17	c.424C>T	p.Arg142Cys	Exon 3	Missense	Huber et al. [1995a]	Compound heterozygous (S42Y/R142C/R323Q)
18	c.425G>A	p.Arg142His[h]	Exon 3	Missense	Russell et al. [1995]	Homozygous
19	c.425G>C	p.Arg142Pro	Exon 3	Missense	Cserhalmi-Friedman et al. [2001]	Compound heterozygous (R142P/ Q582X)
20	c.427C>T	p.Arg143Cys[i]	Exon 3	Missense	Laiho et al. [1997]	Homozygous Compound heterozygous (R143C/V379L) (R142H/R143C)
21	c.428G>A	p.Arg143His[j]	Exon 3	Missense	Russell et al. [1995]	Homozygous
22	c.430G>A	p.Gly144Arg	Exon 3	Missense	Hennies et al. [1998b]	Compound heterozygous (G144R/c.877-2A>G)
23	c.431G>A	p.Gly144Glu[k]	Exon 3	Missense	Petit et al. [1997]	Homozygous
24	c.479C>G	p.Ser160Cys	Exon 3	Missense	Cserhalmi-Friedman et al. [2001]	Heterozygous
25	c.566dupG	p.Ser190GlnfsX49	Exon 4	Frameshift	Farasat et al. [2008]	Compound heterozygous (c.566dupG/c.877-2A>G)
26	c.579G>A	p.Trp193X	Exon 4	Nonsense	Farasat et al. [2008]	Compound heterozygous (W193X/N330H)
27	c.614T>A	p.Leu205Gln[l]	Exon 4	Missense	Akiyama et al. [2001b]	Compound heterozygous (L205Q/R307W)
28	c.625G>T	p.Val209Phe	Exon 4	Missense	This report	Compound heterozygous (R143C/V209F)
29	c.652G>A	p.Gly218Ser[m]	Exon 4	Missense	Laiho et al. [1997]	Compound heterozygous (G218S/R395L)

Number	Nucleotide change[a] NM_000359.2	Amino acid change[b] NP_000350	Exon/ intron	Mutation type	References	Alleles[c]
30	c.674G>A	p.Arg225His	Exon 4	Missense	Hennies et al. [1998b]	Compound heterozygous (R225H/G382R)
31	c.674G>C	p.Arg225Pro	Exon 4	Missense	This report	Compound heterozygous (R225P/c.877-2A>G)
32	c.679C>T	p.Gln227X	Exon 4	Nonsense	Farasat et al. [2008]	Homozygous
33	c.704T>A	p.Leu235X	Exon 4	Nonsense	Farasat et al. [2008]	Compound heterozygous (R126H/L235X)
34	c.728T>G	p.Ile243Ser	Exon 4	Missense	Farasat et al. [2008]	Compound heterozygous (I243S/R315H)
35	c.746C>T	p.Pro249Leu[n]	Exon 4	Missense	Tok et al. [1999]	Compound heterozygous (P249L/F400V)
36	c.788G>A	p.Trp263X	Exon 5	Nonsense	Hennies et al. [1998a]	Homozygous
37	c.790C>T	p.Arg264Trp	Exon 5	Missense	Oji et al. [2006]	Compound heterozygous (R264Q/c.877-2A>G)
38	c.791G>A	p.Arg264Gln	Exon 5	Missense	Oji et al. [2006]	Compound heterozygous (R264Q/c.1074delC)
39	c.802delG	p.Val268PhefsX63	Exon 5	Frameshift	This report	Compound heterozygous (c.802delG/R323Q)
40	c.814T>C	p.Ser272Pro	Exon 5	Missense	Esposito et al. [2001]	Compound heterozygous (S272P/c.877-2A>G)
41	c.826T>A	p.Tyr276Asn	Exon 5	Missense	Oji et al. [2006]	Homozygous
42	c.832G>C	p.Gly278Arg	Exon 5	Missense	Cserhalmi-Friedman et al. [2001]	Compound heterozygous (G278R/R286Q)
43	c.853G>A	p.Glu285Lys	Exon 5	Missense	This report	Compound heterozygous (E285K/A560G)
44	c.857G>A	p.Arg286Gln	Exon 5	Missense	Cserhalmi-Friedman et al. [2001]	Compound heterozygous (G278R/R286Q)
45	c.866A>C	p.Asn289Thr[o]	Exon 5	Misssense	Yang et al. [2001]	Compound heterozygous (N289T/R307W)
46	c.872G>A	p.Gly291Asp	Exon 5	Missense	Farasat et al. [2008]	Compound heterozygous (G291D/R315H)
47	c.876+2T>C	ND	Intron 5	Splice Site	Farasat et al. [2008]	Compound heterozygous (c.876+2T>C/c.877-2A>G)
48	c.877-2A>G[p]	p.Phe293SerfsX38 or p.Phe293ValfsX2	Intron 5	Splice Site	Huber et al. [1995a]	Homozygous
49	c.877T>G	p.Phe293Val	Exon 6	Missense	This report	Compound heterozygous (c.877-2A>G/F293V)
50	c.882_888del CCACGGG	p.Asp294GlufsX34	Exon 6	Frameshift	Farasat et al. [2008]	Compound heterozygous (c.882_888delCCACGGG/R389P)
51	c.910A>T	p.Ile304Phe	Exon 6	Missense	This report	Homozygous
52	c.919C>T	p.Arg307Trp[q]	Exon 6	Missense	Akiyama et al. [2001b]	Compound heterozygous (L205Q/R307W)
53	c.919C>G	p.Arg307Gly	Exon 6	Missense	Oji et al. [2006]	Compound heterozygous (c.877-2A>G/R307G) (R307G/R389P)
54	c.932dupC	p.Tyr312IlefsX23	Exon 6	Frameshift	Farasat et al. [2008]	Compound heterozygous (c.408_411dupCGAC/c.932dupC)
55	c.943C>T	p.Arg315Cys	Exon 6	Missense	Huber et al. [1997]	Compound heterozygous (c.877-2A>G/R315C)
56	c.944G>T	p.Arg315Leu	Exon 6	Missense	Tok et al. [1999]	Homozygous
57	c.944G>A	p.Arg315His	Exon 6	Missense	Oji et al. [2006]	Homozygous
58	c.967C>T	p.Arg323Trp	Exon 6	Missense	This report	Compound heterozygous (c.877-2A>G/R323W) Heterozygous Two patients
59	c.968G>A	p.Arg323Gln	Exon 6	Missense	Huber et al. [1995a]	Compound heterozygous (S42Y/R142C/R323Q)
60	c.984+1G>A	ND	Intron 6	Splice Site	This report	Compound heterozygous with (c.882_888delCCACGGG/ c.984+1G>A/E520G)

Number	Nucleotide change[a] NM_000359.2	Amino acid change[b] NP_000350	Exon/ intron	Mutation type	References	Alleles[c]
61	c.988A>C	p.Asn330His	Exon 7	Missense	Farasat et al. [2008]	Compound heterozygous (W193X/N330H)
62	c.991T>C	p.Ser331Pro	Exon 7	Missense	Farasat et al. [2008]	Compound heterozygous (S331P/c.1420_1421insA)
63	c.1024T>C	p.Trp342Arg	Exon 7	Missense	This report	Compound heterozygous (c.877-2A>G/W342R)
64	c.1042C>T	p.Arg348X	Exon 7	Nonsense	Kon et al. [2003]	Compound heterozygous (R348X/Y365D)
65	c.1074C>G	p.Ser358Arg	Exon 7	Missense	Huber et al. [1997]	Compound heterozygous (S358R/c.2088+1G>T) (S358R/V379L)
66	c.1074delC	p.Ser358ArgfsX26	Exon 7	Frameshift	Oji et al. [2006]	Compound heterozygous (R264Q/c.1074delC)
67	c.1075G>A	p.Val359Met	Exon 7	Missense	This report	Compound heterozygous (V359M/G291D)
68	c.1093T>G	p.Tyr365Asp	Exon 7	Missense	Kon et al. [2003]	Compound heterozygous (R348X/Y365D)
69	c.1097T>C	p.Leu366Pro	Exon 7	Missense	This report	Compound heterozygous (G291D/L366P)
70	c.1107dupA	p.Tyr370IlefsX61	Exon 7	Frameshift	Farasat et al. [2008]	Compound heterozygous (c.877-2A>G/c.1107dupA)
71	c.1135G>C	p.Val379Leu	Exon 7	Missense	Huber et al. [1997]	Compound heterozygous (S358R/V379L)
72	c.1144G>A	p.Gly382Arg or p.Gly382Ser	Exon 7	Missense	Hennies et al. [1998b]	Compound heterozygous (R225H/G382R)
73	c.1147G>A	p.Val383Met[r]	Exon 7	Missense	Petit et al. [1997]	Homozygous
74	c.1159+1G>T	ND	Intron 7	Splice Site	This report	Homozygous
75	c.1166G>C	p.Arg389Pro	Exon 8	Missense	Shevchenko et al. [2000]	Compound heterozygous (c.877-2A>G/R389)
76	c.1166G>A	p.Arg389His[s]	Exon 8	Missense	Akiyama et al. [2001a]	Compound heterozygous (R389H/c.2114delA)
77	c.1175G>A	p.Gly392Asp	Exon 8	Missense	Cserhalmi-Friedman et al. [2001]	Heterozygous
78	c.1186C>A	p.Arg396Ser	Exon 8	Missense	Mizrachi-Koren et al. [2006]	Homozygous
79	c.1187G>T	p.Arg396Leu[t]	Exon 8	Missense	Laiho et al. [1997]	Compound heterozygous (G218S/R396L) (V379L/R396L) Heterozygous
80	c.1187G>A	p.Arg396His	Exon 8	Missense	Farasat et al. [2008]	Compound heterozygous (Y134C/R396H)
81	c.1201T>G	p.Phe401Val[u]	Exon 8	Missense	Tok et al. [1999]	Compound heterozygous (P249L/F401V)
82	c.1213C>A	p.His405Asn	Exon 8	Missense	This report	Compound heterozygous (R389P/H405N)
83	c.1223_1227delACACA	p.Asp408ValfsX21	Exon 8	Frameshift	This report	Homozygous Compound heterozygous (R106X / c.1223_1227delACACA)
84	c.1225_1226delAC[v]	p.Thr409IlefsX21	Exon 8	Frameshift	Bichakjian et al. [1998]	Compound heterozygous (c.408_411dupCGAC/ c.1225_1226delAC)
85	c.1261A>G	p.Met421Val	Exon 8	Missense	This report	Compound heterozygous (c.877-2A>G/M421V)
86	c.1264A>T	p.Lys422X	Exon 8	Nonsense	Cserhalmi-Friedman et al. [2001]	Compound heterozygous (W263X/K422X)
87	c.1289A>T	p.Asp430Val	Exon 8	Missense	Pigg et al. [2000]	Compound heterozygous (V379L/D430V)
88	c.1297delT[w]	p.Trp433GlyfsX11	Exon 8	Frameshift	Huber et al. [1995a]	Homozygous
89	c.1303T>G	p.Phe435Val	Exon 9	Missense	This report	Homozygous
90	c.1313G>A	p.Trp438X	Exon 9	Nonsense	Rice et al. [2005]	Heterozygous

Number	Nucleotide change[a] NM_000359.2	Amino acid change[b] NP_000350	Exon/ intron	Mutation type	References	Alleles[c]
91	c.1331dupA[x]	p.Arg445GlufsX9	Exon 9	Frameshift	Farasat et al. [2008]	Compound heterozygous (Y134C/c.1331dupA)
92	c.1417G>A	p.Gly473Ser	Exon 10	Missense	Huber et al. [1997]	Compound heterozygous (G473S/R687C)
93	c.1420_1421insA[y]	p.Pro474HisfsX24	Exon 10	Frameshift	Farasat et al. [2008]	Compound heterozygous (S331P/c.1420_1421insA)
94	c.1469A>G	p.Asp490Gly	Exon 10	Missense	Raghunath et al. [2003]	Compound heterozygous (G278R/D490G)
95	c.1512C>G	p.Tyr504X[z]	Exon 11	Nonsense	Becker et al. [2003]	Compound heterozygous (Y504X/S670X)
96	c.1555G>T	p.Glu519X	Exon 11	Nonsense	Esposito et al. [2007]	Homozygous
97	c.1559A>G	p.Glu520Gly	Exon 11	Missense	Esposito et al. [2001]	Homozygous
98	c.1631A>G	p.Tyr544Cys	Exon 11	Missense	Farasat et al.[2008]	Homozygous
99	c.1645+1G>A	ND	Intron11	Splice Site	Farasat et al. [2008]	Heterozygous
100	c.1679C>G	p.Ala560Gly	Exon 12	Missense	This report	Compound heterozygous (E285K/A560G)
101	c.1744C>T	p.Gln582X	Exon 12	Nonsense	Cserhalmi-Friedman et al. [2001]	Compound heterozygous (R142P/Q582X)
102	c.1984C>T	p.Gln662X[ab]	Exon 13	Nonsense	Jessen et al. [2000]	Homozygous
103	c.2009C>G	p.Ser670X[ac]	Exon 13	Nonsense	Becker et al. [2003]	Compound heterozygous (Y504X/S670X)
104	c.2059delC[ad]	p.Arg687ValfsX64	Exon 13	Frameshift	Lugassy et al. [2007]	Homozygous
105	c.2059C>T	p.Arg687Cys	Exon 13	Missense	Huber et al. [1997]	Compound heterozygous (G473S/R687C)
106	c.2060G>A	p.Arg687His	Exon 13	Missense	Oji et al. [2006]	Homozygous
107	c.2065C>T	p.Arg689Cys	Exon 13	Missense	This report	Heterozygous
108	c.2066G>A	p.Arg689His	Exon 13	Missense	This report	Compound heterozygous (R348X/R689H)
109	c.2088+1G>T	p.Val679_Thr696del	Intron 13	Splice Site	Huber et al. [1997]	Compound heterozygous (S358R/c.2088+1G>T)
110	c.2090T>G	p.Leu697X	Exon 14	Nonsense	Farasat et al. [2008]	Compound heterozygous (R315H/L687X)
111	c.2114delA[ae]	p.Gln705ArgfsX46	Exon 14	Frameshift	Akiyama et al. [2001a]	Compound heterozygous (R389H/c.2114delA)
112	c.2226-2A>G	ND	Intron 14	Splice Site	This report	Compound heterozygous (c.877-2A>G/ c.2226-2A>G)
113	c.2278C>T	p.Arg760X	Exon 15	Nonsense	Shevchenko et al. [2000]	Compound heterozygous (c.877-2A>G/R760X)
114	c.2290C>T	p.Arg764Cys	Exon 15	Missense	Mizrachi-Koren et al. [2006]	Homozygous
115	c.2320C>T	p.Gln774X	Exon 15	Nonsense	Esposito et al. [2007]	Homozygous

***TGM1* Polymorphisms**

Number	Nucleotide change[a] NM_000359.2	Amino acid change[b]	Exon/ intron	References
1	c.726G>A[af]	NA	Exon 4	Huber et al. [1995b]
2	c.877-3C>G[ag]	NA	Intron 5	Huber et al. [1995b]
3	c.1146C>A	NA	Exon 7	Hennies et al. [1998b] and this report
4	c.1552G>A	p.Val518Met	Exon 11	Hennies et al. [1998b]

[a]The nucleotide numbering is based on the published GenBank sequence NM_000359.2, and nucleotide +1 is the adenine of the ATG initiation codon.
[b]The amino acid sequence is taken from the published GenBank sequence NP_000350 and begins with the translation initiation codon as 1.
[c]*TGM1* alleles of affected patients in the first report of each mutation. Alleles and number of patients are indicated.
[d]Mutation was reported as p.D101V in Yang et al. [2001].
[e]Mutation was reported as c.343insT in Farasat et al. [2008].
[f]Mutation was reported as c.371delA in Akiyama et al. [2003].
[g]Mutation was reported as 1387insCAGC in Bichakjian et al. [1998]
[h]Mutation was reported as p.Arg141His in Russell et al. [1995].
[i]Mutation was reported as p.Arg142Cys in Laiho et al. [1997].
[j]Mutation was reported as p.Arg142His in Russell et al. [1995].
[k]Mutation was reported as p.Gly143Glu in Petit et al. [1997].
[l]Mutation was reported as p.Leu204Gln in Akiyama et al. [2001b].
[m]Mutation was reported as p.Gly217Ser in Laiho et al. [1997].
[n]Mutation was reported as p.Pro248Leu in Tok et al. [1999].
[o]Mutation was reported as p.Asn288Thr in Yang et al. [2001].
[p]Mutation was reported as +3366A>G in Huber et al. [1995].
[q]Mutation was reported as p.Arg306Trp in Akiyama et al. [2001b].
[r]Mutation was reported as p.Val382Met in Petit et al. [1997].
[s]Mutation was reported as p.Arg388His in Akiyama et al. [2001a].
[t]Mutation was reported as p.Arg395Leu in Laiho et al. [1997].
[u]Mutation was reported as p.Phe400Val in Tok et al. [1999].
[v]Mutation was reported as 4561delAC in Bichakjian et al. [1998].
[w]Mutation was reported as +4640delT in Huber et al. [1995].
[x]Mutation was reported as c.1331insA in Farasat et al. [2008].
[y]Mutation was reported as c.1421insA in Farasat et al. [2008].
[z]Mutation was reported as p.Tyr503X in Becker et al. [2003].
[ab]Mutation was reported as p.Q661X in Jessen et al. [2000].
[ac]Mutation was reported as p.S669X in Becker et al. [2003].
[ad]Mutation was reported as c.2058delC in Lugassay et al. [2007].
[ae]Mutation was reported as 9008delA in Akiyama et al. [2001a].
[af]Polymorphism was reported as +2698 G>A in Huber et al. [1995b].
[ag]Polymorphism was reported as +3365G>C in Huber et al. [1995b].
NA, not applicable ND, not determined

Supp. Table S2. Reported TGase-1 activity in cultured keratinocytes from patient biopsies

Mutation (Patient)	Membrane activity	%	Cytosolic activity	%	Control (Membrane/ Cytosolic)	Domain
Unaffected heterozygote R307W[a] (I-1)	1007.52 ± 104.29	(52)	305.96 ± 110.18	(56)	(NA/ NA)	Catalytic core
Unaffected heterozygote D102V[a] (I-2)	939.12 ± 205.91	(48)	284.07 ± 92.63	(52)	(NA/ NA)	β –sandwich
R307W/D102V[a] (II-2)	102.41 ± 27.28	(5)	82.32 ± 42.12	(15)	(NA/ NA)	Catalytic core/ β-sandwich
Homozygous V383M[b] (B5)	186.2 ± 78.7	(4; 6)	26.5 ± 6.7	(5; 7)	(5282 ± 387/ 547.7 ± 44.9); (2980 ± 592/ 358.9 ± 59.8)	Catalytic core
S358R/ V379L[c] (LI-20 II-1)	175.4 ± 44.0	(4)	76.3 ± 5.5	(22)	(4200.0 ± 200.0/ 351.9 ± 125.7)	Catalytic core/ Catalytic core
Homozygous c.877-2G>A[d] (LI-2.10)	6.5 ± 3.0	(3; 3)	1.1 ± 0.3	(4; 6)	(247 ± 57/ 29.3 ± 2.4); (221 ± 97/ 19.2 ± 3.2)	Intron 5
Homozygous c.877-2G>A[d] (LI-2.9)	6.3 ± 1.0	(3; 3)	0.72 ± 0.1	(2; 4)	(247 ± 57/ 29.3 ± 2.4); (221 ± 97/ 19.2 ± 3.2)	Intron 5
S42Y/R142Cys/ R323Gln[d] (LI-3.7)	5.0 ± 1.0	(2; 2)	1.95 ± 0.4	(7; 10)	(247 ± 57/ 29.3 ± 2.4); (221 ± 97/ 19.2 ± 3.2)	Anchoring/ β-sandwich/ Catalytic core
Homozygous c.1297delT[d] (LI-1.5)	4.0	(2; 2)	0.33	(1; 2)	(247 ± 57/ 29.3 ± 2.4); (221 ± 97/ 19.2 ± 3.2)	Catalytic core
R315C/ c.877-2A>G[c] (LI-22 IV.4)	52.0 ± 4.9	(1)	4.0 ± 2.4	(1)	(4200.0 ± 200.0/ 351.9 ± 125.7)	Catalytic core/ Intron 5
S42Y/R142Cys/ R323Gln[d] (LI-3.8)	1.8 ± 1.0	(0.7; 0.8)	1.90 ± 1.4	(6; 10)	(247 ± 57/ 29.3 ± 2.4); (221 ± 97/ 19.2 ± 3.2)	Anchoring/ β-sandwich/ Catalytic core
G473S/ R687C[c] (LI-11 II.1)	24.3 ± 7.3	(0.6)	10.9 ± 2.2	(3)	(4200.0 ± 200.0/ 351.9 ± 125.7)	Catalytic core/ B-Barrel 1
Homozygous R127X[c] (LI-8 II.2)	13.9 ± 1.8	(0.3)	5.8 ± 1.1	(2)	(4200.0 ± 200.0/ 351.9 ± 125.7)	β –Sandwich
Homozygous G144E[b] (1)	10.1 ± 1.9	(0.2; 0.3)	14.4 ± 9.4	(3; 4)	(5282 ± 387/ 547.7 ± 44.9); (2980 ± 592/ 358.9 ± 59.8)	β –Sandwich
S358R/ c.2088+1G>T[c] (LI-20 III.2)	4.0 ± 1.9	(0.1)	5.6 ± 2.1	(2)	(4200.0 ± 200.0/ 351.9 ± 125.7)	Catalytic core/ β-Barrel 2
S358R/ c.2088+1G>T[c] (LI-20 III.1)	2.2 ± 0.3	(0.05)	2.0 ± 1.1	(0.6)	(4200.0 ± 200.0/ 351.9 ± 125.7)	Catalytic core/ β-Barrel 2

[a]Patients with these mutations diagnosed with LI and reported as mean ± S.E. [Yang et al., 2001]

[b]Patients with these mutations diagnosed with LI and reported as mean ± SEM [Petit et al., 1997]

[c]Patients with these mutations diagnosed with LI and reported as mean ± S.E. [Huber et al., 1997]

[d]Patients with these mutations diagnosed with LI and reported as mean ± S.D. [Huber et al., 1995a]

Units expressed as pmol of ^{3}H –putrescine incorporated into dimethylcaseine per hour per mg of protein, except for those reported by Huber et al., 1995a which are expressed as disintegrations per minute per milligram of protein × 10^-4. The % of control activities are given to the right of both columns reporting the cytosolic and membrane activities. If more than one control is used then a semicolon separates the two percentages relative to the control values as well as the control values themselves. Activity assays were all similar and done by a protocol previously described [Lichti et al., 1985].

Supp. Table S3. Reported TGase-1 Activity in Sf9 cells transfected with a baculovirus vector

Mutation	Membrane	%	Cytosolic	%	Control (Membrane/ Cytosolic)	Domain
R315L[a]	13.0 ± 14.5	222	33.6 ± 10.8	200	(5.9 ± 1.6/ 16.8 ± 6.7)	Catalytic core
S42Y[a]	11.4 ± 3.3	182	74.4 ± 12	442	(5.9 ± 1.6/ 16.8 ± 6.7)	Anchoring
D490G[b,c]	4.7 ± 2.8	85	12.5 ± 2.4	81	(5.5 ± 2.0/ 15.5 ± 5.6)	Catalytic core
R323Q[a]	1.5 ± 1.2	26	12.2 ± 2.4	72	(5.9 ± 1.6/ 16.8 ± 6.7)	Catalytic core
R143H[a]	<0.01	0	3.4 ± 0.1	20	(5.9 ± 1.6/ 16.8 ± 6.7)	β-Sandwich
R142H[a]	<0.01	0	0.5 ± 0.1	3	(5.9 ± 1.6/ 16.8 ± 6.7)	β-Sandwich
G278R[b,d]	-	0	0.3 ± 0.1	2	(5.5 ± 2.0/ 15.5 ± 5.6)	Catalytic core
R142C[a]	<0.01	0	<0.01	0	(5.9 ± 1.6/ 16.8 ± 6.7)	β-Sandwich
c.1297del T[a]	<0.01	0	<0.01	0	(5.9 ± 1.6/ 16.8 ± 6.7)	Catalytic core

[a]Patients with these mutations diagnosed with LI and reported as mean ± S.D. [Candi et al., 1998].
[b]Patients with these mutations diagnosed with SHCB; analysis done under ambient pressure [Raghunath et al., 2003].
[c]Under increased ambient pressure, +115mmHg, the protein had the following activities: membrane 4.7 ± 0.9 (39%), cytosolic 0.8 ± 0.5 (22%), control 12.2 ± 2.5/ 3.7 ± 1.1 (100%) [Raghunath et al., 2003].
[d]Under increased ambient pressure, +115mmHg, the protein had the following activities: membrane 0.2 ± 0.2 (2%), cytosolic – (0%), control 12.2 ± 2.5/ 3.7 ± 1.1 (100%) [Raghunath et al., 2003].
Units expressed as pmol of [^{14}C] putrescine incorporated into succinylated casein/h/pmol of TGase 1. The % of control activities are given to the right of both columns reporting the cytosolic and membrane activities. If more than one control is used then a "–" separates the two percentages relative to the control values as well as the control values themselves. The specific activity protocols were all similar and done by a protocol previously described [Candi et al., 2003].