

Supplementary table S3: Summary of Immunohistochemistry Findings

Patient No.	Age (months)/Sex	Blister Cavity	Immunohistochemistry					Mutational Analysis	
			Keratin 14 (NCL-LL0002)	Laminin-332 (P3H9-2)	Collagen IV (CIV-22)	Collagen VII (NCL-COLL-VII)	Diagnosis	Gene	Diagnosis
1	42; Male	Present	Near Normal; Both sides	Normal; Floor	Normal; Floor	Normal; Floor	EBS	KRT5	EBS
2	18; Male	Present	Absent	Normal; Floor	Normal; Floor	Normal; Floor	EBS	KRT14	EBS
3	108; Female	Present	Absent	Normal; Floor	Normal; Floor	Normal; Floor	EBS	KRT14	EBS
4	0.13; Female	Present	Absent	Normal; Floor	Normal; Floor	Normal; Floor	EBS	KRT14	EBS
5	216; Male	Present	Normal; Roof	Near Normal; Floor	Normal; Floor	Normal; Floor	JEB	LAMB3	JEB
6	60; Male	Present	Normal; Roof	Faint; Floor	Normal; Floor	Normal; Floor	JEB	ITGB4	JEB
7	0.03; Male	Present	Normal; Roof	Faint; Floor	Normal; Floor	Normal; Floor	JEB	ITGB4	JEB
8	192; Female	Present	Normal; Roof	Near Normal; Floor	Normal; Floor	Normal; Floor	JEB	LAMB3	JEB
9	96; Male	Present	Normal; Roof	Near Normal; Floor	Normal; Floor	Normal; Floor	JEB	LAMB3	JEB
10	108; Male	Present	Normal; Roof	Near Normal; Floor	Normal; Floor	Normal; Floor	JEB	LAMA3	JEB
11	3; Male	Present	Normal; Roof	Absent	Normal; Floor	Normal; Floor	JEB	LAMB3	JEB
12	6; Female	Present	Near Normal; Roof	Normal; Floor	Normal; Floor	Normal; Floor	EBS	COL7A1	DEB
13	12; Female	Present	Absent	Absent	Reduced; Floor	Absent	Inconclusive	COL7A1	DEB
14	48; Female	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	COL7A1	DEB
15	6; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	COL7A1	DEB
16	288; Male	Present	Normal; Roof	Normal; Both Sides	Reduced; Both Sides	Faint; Both Sides	DEB	COL7A1	DEB
17	24; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	COL7A1	DEB
18	144; Male	Present	Near Normal; Roof	Normal; Floor	Normal; Floor	Normal; Floor	EBS	COL7A1	DEB
19	36; Female	Present	Near Normal; Roof	Normal; Floor	Normal; Floor	Normal; Both Sides	EBS	COL7A1	DEB
20	26; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	COL7A1	DEB
21	48; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	COL7A1	DEB

Supplementary table S3: Summary of Immunohistochemistry Findings (contd.)

Patient No.	Age (months)/Sex	Blister Cavity	Immunohistochemistry					Mutational Analysis	
			Keratin 14 (NCL-LL0002)	Laminin-332 (P3H9-2)	Collagen IV (CIV-22)	Collagen VII (NCL-COLL-VII)	Diagnosis	Gene	Diagnosis
22	48; Female	Present	Absent	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
23	360; Male	Present	Normal	Absent	Reduced; Roof	Absent	Inconclusive	Unknown	Unknown
24	12; Male	Present	Near Normal; Both sides	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
25	276; Female	Present	Near Normal; Both sides	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
26	36; Female	Present	Normal; Both sides	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
27	60; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	Unknown	Unknown
28	120; Male	Present	Normal; Roof	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
29	12; Female	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	Unknown	Unknown
30	0.67; Female	Present	Normal; Roof	Near Normal; Floor	Normal; Floor	Normal; Floor	JEB	Unknown	Unknown
31	72; Male	Present	Near Normal; Both sides	Normal; Floor	Normal; Floor	Normal; Floor	EBS	Unknown	Unknown
32	168; Male	Present	Normal; Roof	Normal; Roof	Normal; Roof	Absent	DEB	Unknown	Unknown
33	24; Female	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL17A1	JEB
34	18; Male	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
35	84; Male	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
36	168; Male	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
37	24; Male	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
38	108; Male	No Split	Normal	Normal	Normal	Reduced	DEB	COL7A1	DEB
39	2; Female	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
40	24; Male	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
41	60; Female	No Split	Normal	Normal	Normal	Normal	Inconclusive	COL7A1	DEB
42	18; Female	No Split	Normal	Normal	Normal	Normal	Inconclusive	Unknown	Unknown
43	420; Female	No Split	Normal	Normal	Normal	Normal	Inconclusive	Unknown	Unknown
44	48; Male	No Split	Normal	Absent	Absent	Absent	Inconclusive	Unknown	Unknown