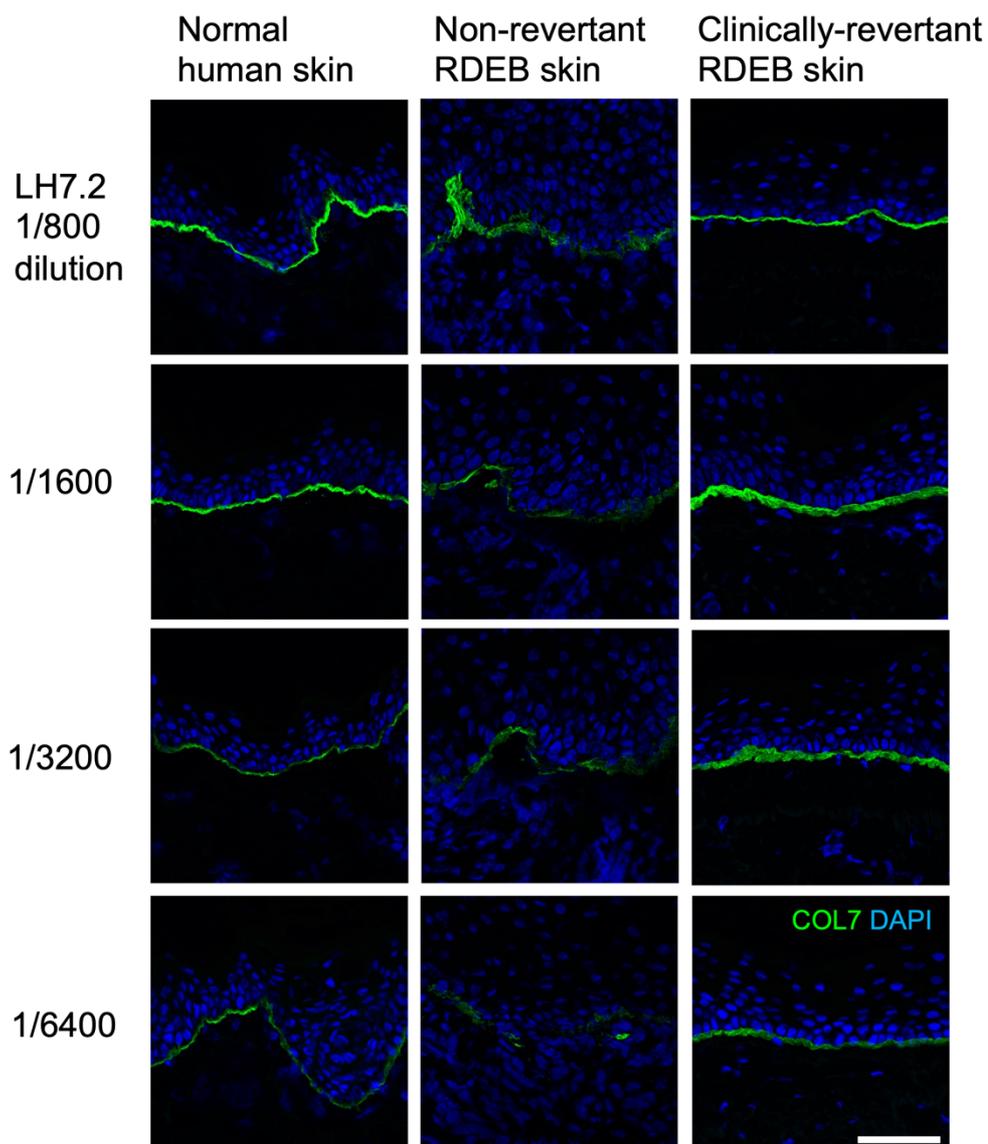




Title	Detection of revertant mosaicism in epidermolysis bullosa through Cas9-targeted long-read sequencing
Author(s)	Natsuga, Ken; Furuta, Yoshikazu; Takashima, Shota et al.
Citation	Human mutation, 43(4), 529-536 https://doi.org/10.1002/humu.24331
Issue Date	2022-04-01
Doc URL	https://hdl.handle.net/2115/88789
Rights	This is the peer reviewed version of the following article: Natsuga, K., Furuta, Y., Takashima, S., Nohara, T., Kosumi, H., Mai, Y., Higashi, H., & Ujiie, H. (2022). Detection of revertant mosaicism in epidermolysis bullosa through Cas9-targeted long-read sequencing. Human Mutation, 43, 529- 536 which has been published in final form at doi:10.1002/humu.24331. This article may be used for non-commercial purposes in accordance with Wiley Terms and Conditions for Use of Self-Archived Versions.
Type	journal article
File Information	Supplementary_Information_Hum_Mutat_R2.pdf





Supplementary Figure 1. COL7 staining of clinically revertant and non-revertant skin.

Samples of normal human skin, non-revertant RDEB skin, and clinically revertant RDEB skin stained with COL7 (primary antibody LH7.2 dilution: 1/800, 1/1600, 1/3200, 1/6400). Bar = 100 μ m.

Supplementary Table 1. The gRNA sequences and their target sites.

gRNA name	gRNA sequence	Target site (GRCh38/hg38)
COL7A1_c.5932_fwd1	TGGGGAGACTGGATCCAAAG	Chr3:(-) 48576408
COL7A1_c.5932_fwd2	TCAAGGGAACCTGGGCAGTG	Chr3:(-) 48576082
COL7A1_c.5932_fwd3	GGAAACTGCTGGCATCAAGG	Chr3:(-) 48575866
COL7A1_c.8029_rev1	CCCTCAATTCACCATGACCA	Chr3:(+) 48567094
COL7A1_c.8029_rev2	GGGATCAGGAGTCAGAGCTG	Chr3:(+) 48566876
COL7A1_c.8029_rev3	ACACAGGGACCAAGTGAGCA	Chr3:(+) 48566745

Supplementary Table 2. Read counts of COL7A1 haplotypes by nCATS.

Sample	WT	c.5932C>T	c.8029G>A	Double mutant	Others ^a	Total
Normal human epidermis	34	0	0	0	0	34
Non-revertant dermis	3	70	50	1	5	129
Non-revertant epidermis	1	15	12	2	2	32
Revertant dermis	2	122	101	1	10	236
Revertant epidermis	39	5	4	17	1	66

a. Haplotypes with unexpected bases or indels at the loci.