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Fig. S1. Schematic representation of the BPAG1-e protein. The previously reported *DST* mutations are indicated above the schematic protein structure with blue arrows (the thin blue arrow points to a variant which was considered as probably not decisive for the phenotype). The homozygous p.Ser2388Leufs*5 variant identified in our patient is located in the intermediate filament binding domain (IFBD) and is indicated with a red arrow. Schematic representation modified from Turcan et al (5).

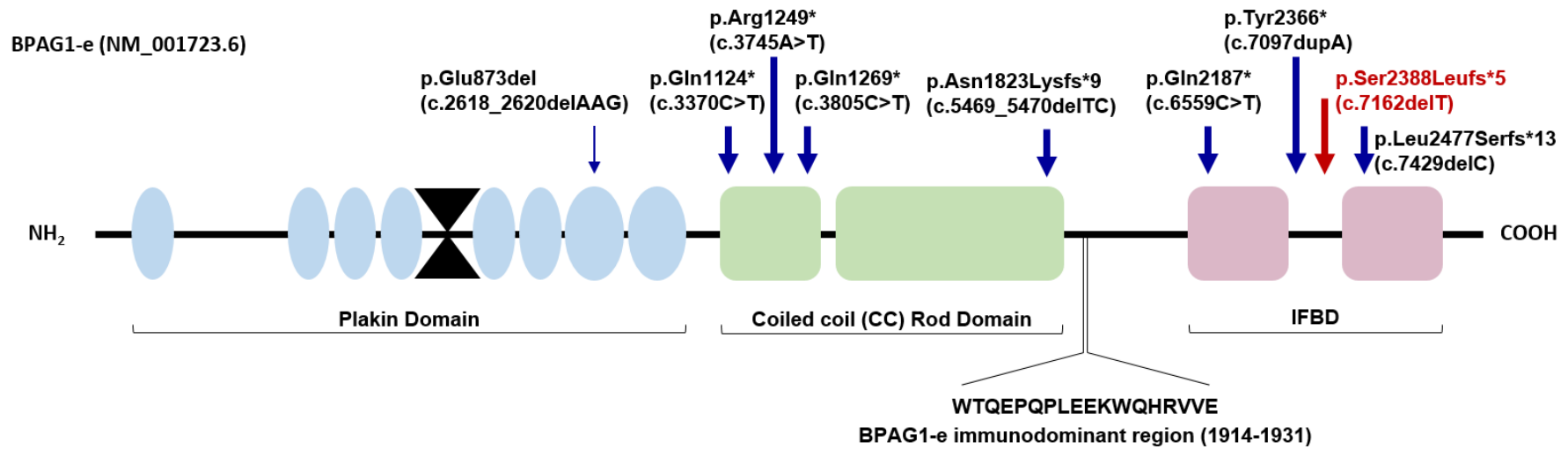


Fig. S2. Immunofluorescence analysis of the patient's skin. Keratin 15 and CD151 staining were decreased compared to control skin. Staining intensities of the other HD components and junctional proteins tested were comparable to the control.

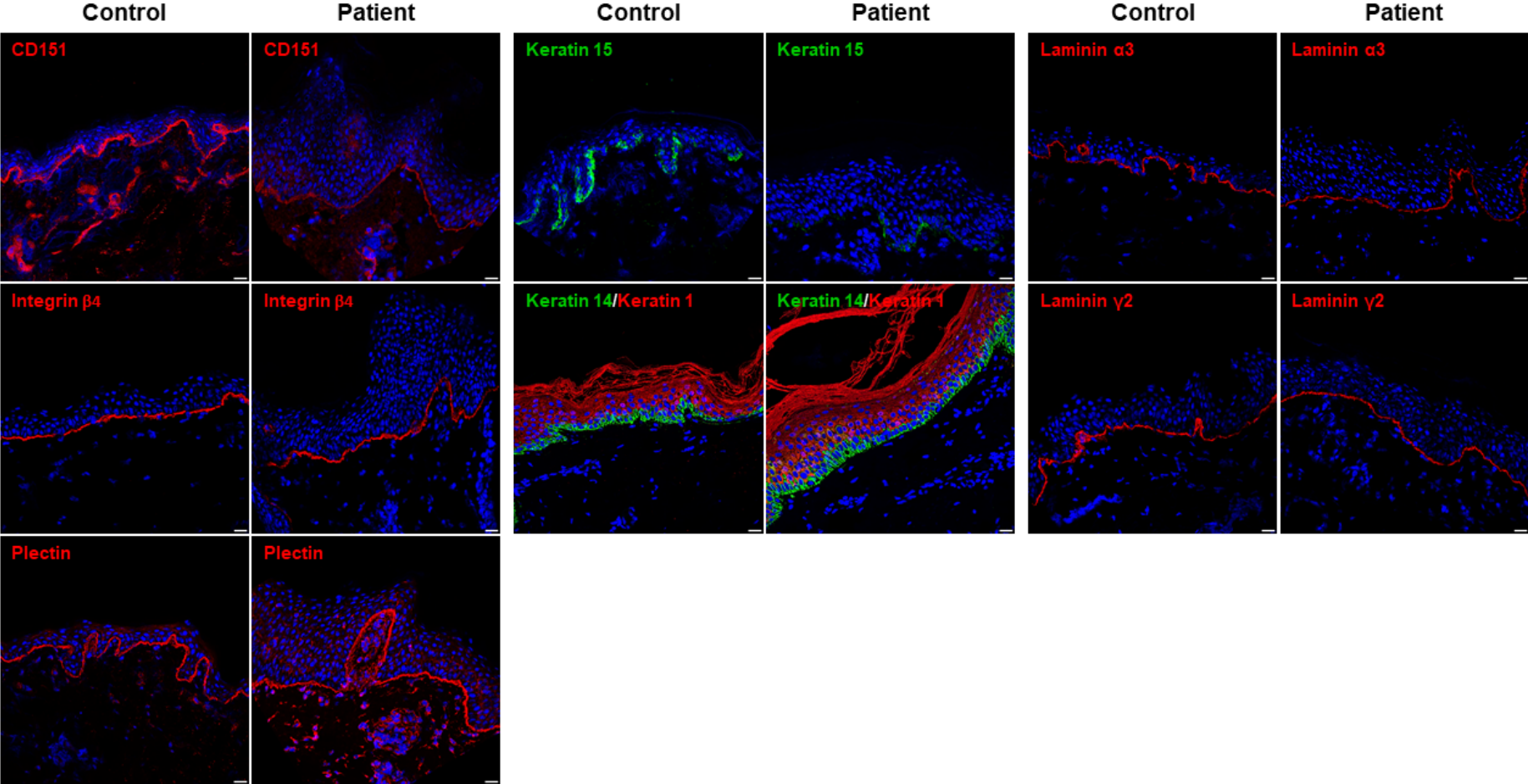


Fig. S3. Representation of the cutaneous dermal-epidermal junction with HD and FA proteins whose expression is altered in the patient. Proteins in red frames represent downregulated proteins or proteins with an abnormal distribution pattern in our patient. Proteins in yellow frames represent proteins with expression levels comparable to those of a healthy control. Proteins in green frames represent upregulated proteins. Schematic representation modified from Bardhan et al (6).

