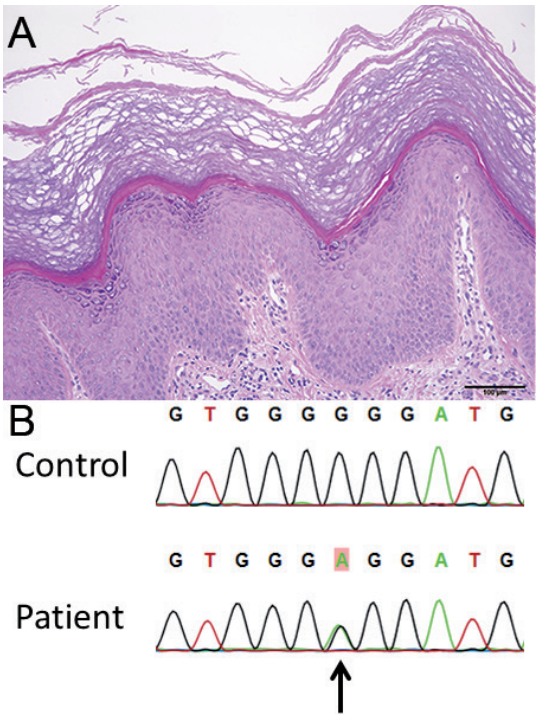

Fig. S1. (A) Haematoxylin-eosin staining shows hyperkeratosis with a slightly thickened granular layer and mild acanthosis. (B) Sanger sequencing reveals a heterozygous mutation within *GJB3*, c.134G>A (p.Gly45Glu) in the proband, but not in control DNA. The original magnification × 200.

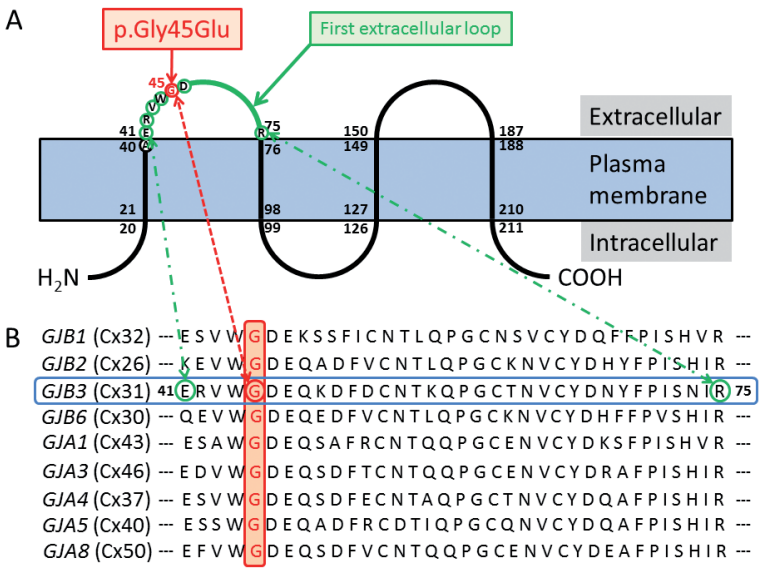

Fig. S2. Position of the first glycine in the first extracellular loop of Cx31. (A) Schematic model of Cx31 structure; the mutation in the proband is marked by a *red arrow*; the first extracellular domain is coloured *green*. The *black* numbers indicate the numbering of the amino acids for Cx31. (B) This corresponding glycine (*red arrow*) is a highly conserved residue among 9 Cx gene family members.

Table SI. *Reported missense mutations of the first glycine in the first extracellular domains of the connexin gene family*

Gene	Protein	Substitution	Type of mutation	Phenotype	Ref.
GJB2	Cx26	p.Gly45Glu	Germline	KID syndrome	6, 7
		p.Gly45Glu	Somatic	PEODDN	8
GJB3	Cx31	p.Gly45Glu	Germline	EKV	4, 5, this case
GJA8	Cx50	p.Gly46Arg	Germline	Congenital cataract and microcornea	11
		p.Gly46Val	Germline	Congenital cataract and microcornea	12

Cx; connexin; KID; keratitis, ichthyosis and deafness; PEODDN: porokeratotic eccrine ostial and dermal duct naevus; EKV: erythrokeratoderma variabilis.