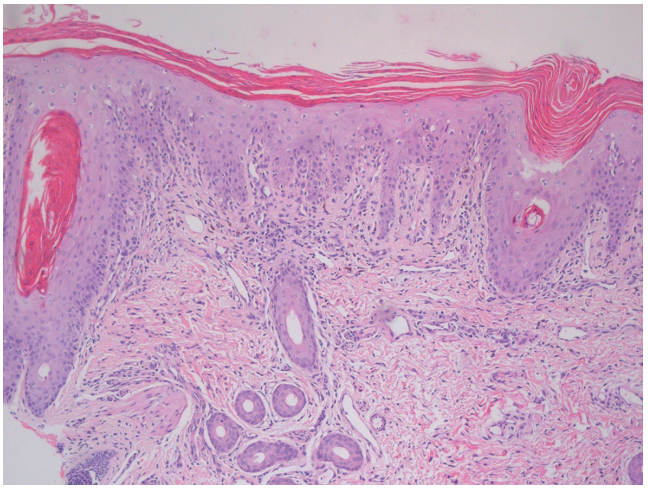

Fig. S1. Mild regular acanthosis, large keratotic plugs located within hair follicles and follicular and perifollicular parakeratosis. In the upper dermis, hyperplastic capillaries surrounded by a relatively dense infiltrate of lymphocytes and neutrophils (HES ×100).

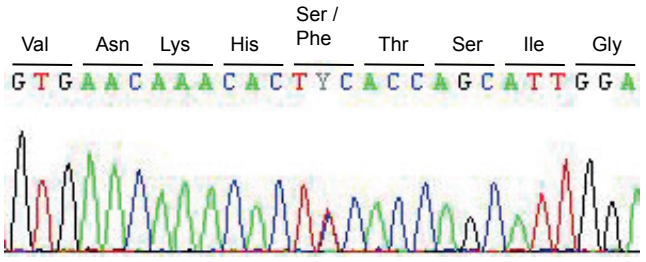

Fig. S2. Identification of a *GJB2* mutation in the patient. Mutation screening of the *GJB2* coding sequence was performed by PCR amplification from position c.-22-170 to position c.*77 (primers to amplify 2 overlapping fragments: forward primers 5'CCTATGACAAACTAAGTTGGTTCTGTCTT3' and 5'CGGAGACATGAGAAGAAGAG3' with respective reverse primers 5'AGCCTTCGATGCGGACCTTC3' and 5'GAGCCTTGACAGCTGAGCAC3') followed by a bidirectional Sanger sequencing. A representative chromatogram showing a heterozygous C >T transition at position c.50 corresponding to a substitution of the normal serine codon to a phenylalanine codon (p.Ser17Phe), is shown. Nomenclature of the mutation refers to the *GJB2* RefSeq NM_00400.5, with nucleotide number +1 beginning at the A of the start codon ATG.

Table SI. *Characteristics of the 9 families (12 patients) reported in the literature and our patient*

Reference	3	6	7	8, 9	10	11	12	13	14	Our patient
Familial cases	–	2 dizygotic twins, 1 sibling	–	–	2 dizygotic twins	–	–	–	–	–
Origin	ND	African	Caucasian	Caucasian	African	ND	ND	Asian	ND	African
Suspected mode of inheritance	*De novo*	Germline, mosaic	*De novo*	*De novo*	Somatic, mosaic	*De novo*	*De novo*	*De novo*	*De novo*	*De novo*
Age at first signs	Birth	Birth	2 months	Birth	Birth	Birth	Birth	Birth	Birth	1 Month
Dystrophic nails	+	+	–	+	+	–	–	+	+	+
Keratitis	–	–	+	+	–	–	–	+	+	+
Hearing deficit	+	ND	+	+	ND	ND	+	+	+	ND
Other abnormalities	+	+	–	–	+	+	+	–	–	–
Age at death	3 months	1 month, 1 month, 5 months	12 months	6 months	30 days, 5 months	2 months	3 months	51 days	77 days	15 months
Cause of death	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia
GJB2 mutation	p.Ala88Val	p.Gly45Glu	p.Gly45Glu	p.Gly45Glu	p.Gly45Glu	ND	ND	ND	ND	p.Ser17Phe

ND: not determined.