Supplementary material to article by J. Mazereeuw-Hautier et al. “Lethal Form of Keratitis–Ichthyosis–Deafness Syndrome Caused by the GJB2 Mutation p.Ser17Phe”

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’CCTATGACAAACTAAGTTGGTTCTGTTCTT3’ and 5’CGGAGACATGAGAAGAAGG3’ 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.