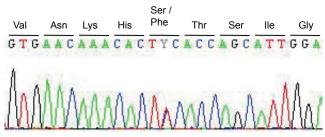
Supplementary material to article by J. Mazereeuw-Hautier et al. "Lethal Form of Keratitis–Ichthyosis–Deafness Syndrome Caused by the GJB2 Mutation p.Ser17Phe"



Heterozygous *GJB2* mutation: c.50C>T, 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'CCTATGACAAACTAAGTTGGTTCTGTCTT3' and 5'CGGAGACATGAGAAGAAGAAGAAGAG3' with respective reverse primers 5'AGCCTTCGATGCGGACCTTC3' and 5'GAGCCTTGACAAGCTGAGCAC3') 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.