Supplementary material to article by P. Kirchmeier et al. "Whole-Exome-Sequencing Reveals Small Deletions in CASP14 in Patients with Autosomal Recessive Inherited Ichthyosis"



Fig. S1. Patients' pedigrees, wild type and mutated CASP14 protein and sequencing results of family A. (a) Family A. The father of patients 1 and 2 married a first cousin once removed. (b) Family B. Both unaffected siblings present the wild-type sequence. Alleles are shown in parentheses. (c) Diagram of the wild-type and the hypothetical mutated CASP14 protein structure. The wild-type protein consists of a short pro-domain (yellow), the large catalytic subunit p17 (blue) and the small subunit p11 (green). The hypothetical protein structure illustrates the amino acid sequence alteration (aa 154) and the truncation of the subunit p11 (aa 180; red). (d) The healthy parents are heterozygous, while the patients are homozygous for the c.462_463delCA deletion (CASP14, NM_012114).

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