



Fig. S1. ALDH3A2 mutations in the patient with Sjögren-Larsson syndrome and sequence alignments around the missense mutation. (A) Direct sequencing reveals a heterozygous c.1157A>G (p.N386S) transition in exon 8 of the *ALDH3A2* gene in the patients and their mother, but not in their father or normal control samples. (B) A heterozygous c.1291\_1292delAA (p.Lys431Glufs\*5) mutation is found in exon 9 of *ALDH3A2* in the patients and their father, but not in their mother or in normal controls. (C) Fatty aldehyde dehydrogenase (FALDH) amino-acid sequence alignment shows the level of conservation in diverse species of the amino-acid N386 (red characters), which was altered by the missense mutation in the present family. (D) A sequence alignment between the FALDH, rat class 3 and human class 1 and class 2 ALDHs showing the relative locations of key residues in these enzymes. The asparagine residue at codon 386 of FALDH is strictly conserved (red characters). Secondary structure components found in the class 3 rat ALDH structure are indicated with a bar and an arrow. The bar represents the  $\alpha$ -helix and arrow represents  $\beta$ -strands. (Modified from Liu et al. (7)).