Supplementary Figure 1. SLC29A3 homozygous mutation in the patient

A. Sequence of the patient and control genomic DNA, showing a homozygous mutation in the splice site of intron 2 of the SLC29A3 gene, located at c.300+1G>C on SLC29A3 cDNA (NM_018344). Reference genomic sequence is shown above the sequence tracks (exon: capital letters, intron: lower case).

B. Predicted consequence of the mutation, resulting in a truncated protein (N101LfsX34 on NP_060814).

Supplementary Figure 1 legend
A. Sequence of the patient and control genomic DNA, showing a homozygous mutation in the splice site of intron 2 of the SLC29A3 gene, located at c.300+1G>C on SLC29A3 cDNA (NM_018344). Reference genomic sequence is shown above the sequence tracks (exon: capital letters, intron: lower case). B. Predicted consequence of the mutation, resulting in a truncated protein (N101LfsX34 on NP_060814).