Appendix S1

SUPPLEMENTARY MATERIAL

Genomic DNA was extracted from the blood of the patient, and a healthy control. NucleoSpin® Blood (Takara, Shiga, Japan) was used for DNA extraction, according to the manufacturer's instructions.

Total RNA was extracted from the blood of healthy control and patient. TRIzol™ Reagent (Invitrogen, Waltham, MA, USA) and Direct-zol™ RNA MiniPrep (Zymo Research, Irvine, CA, USA) were used for RNA extraction, according to the manufacturer's instructions.

In order to perform a sequence analysis of *TMC6*, genomic DNA or complemental DNA (cDNA) was used as a template and amplified by PCR using KOD One PCR Master Mix -Blue- (TOYOBO, Osaka, Japan). The primers for *TMC6* used in this study are shown in Table SI. Primer sequences for the other coding region are available from the authors on request. The PCR product was electrophoresed on a 2.0% agarose gel and gel recovered using the Wizard® SV Gel and PCR Clean-Up System (Promega, Madison, WI, USA). The recovered PCR products were treated with the BigDye® Direct Cycle Sequencing Kit (Applied Biosystems, Waltham, MA, USA), subjected to ethanol precipitation and sequence analysis (Applied Biosystems 3730 DNA Analyzer). The sequence data were analysed using ApE (https://jorgensen.biology.utah.edu/wayned/ape/) and FinchTV (https://digitalworldbiology.com/finchtv).