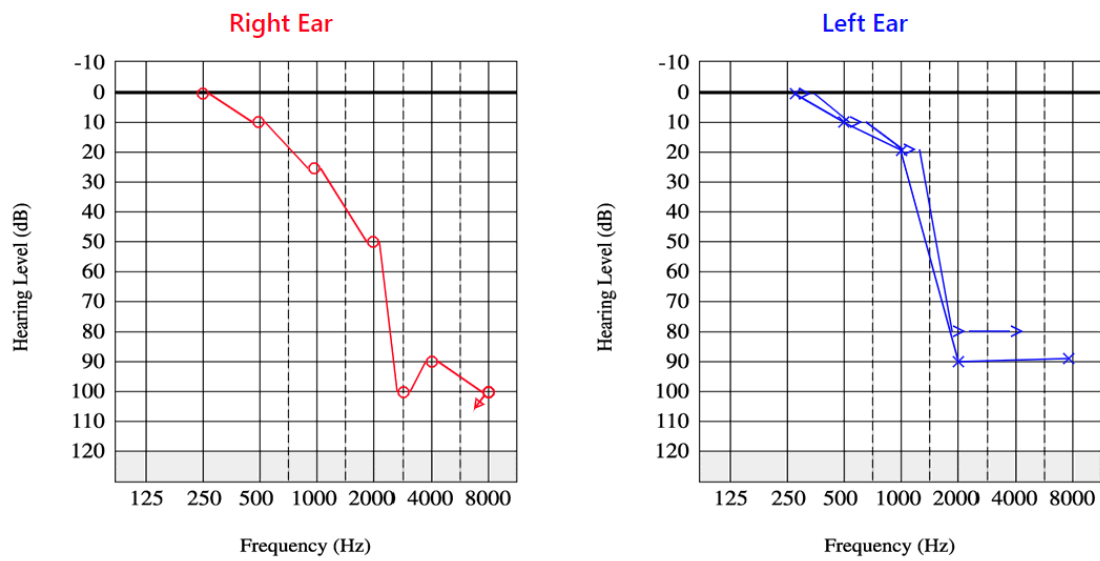
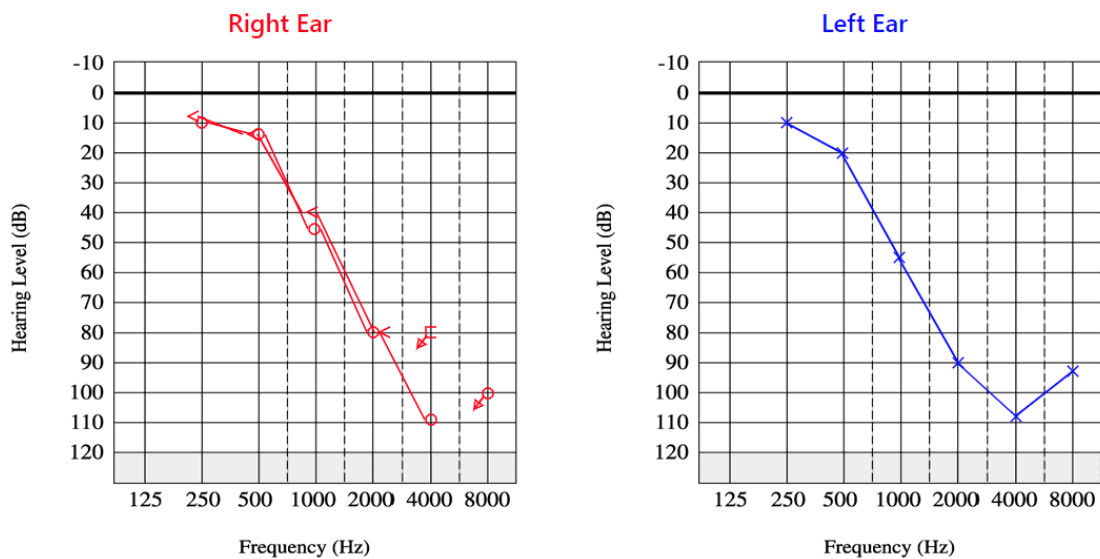


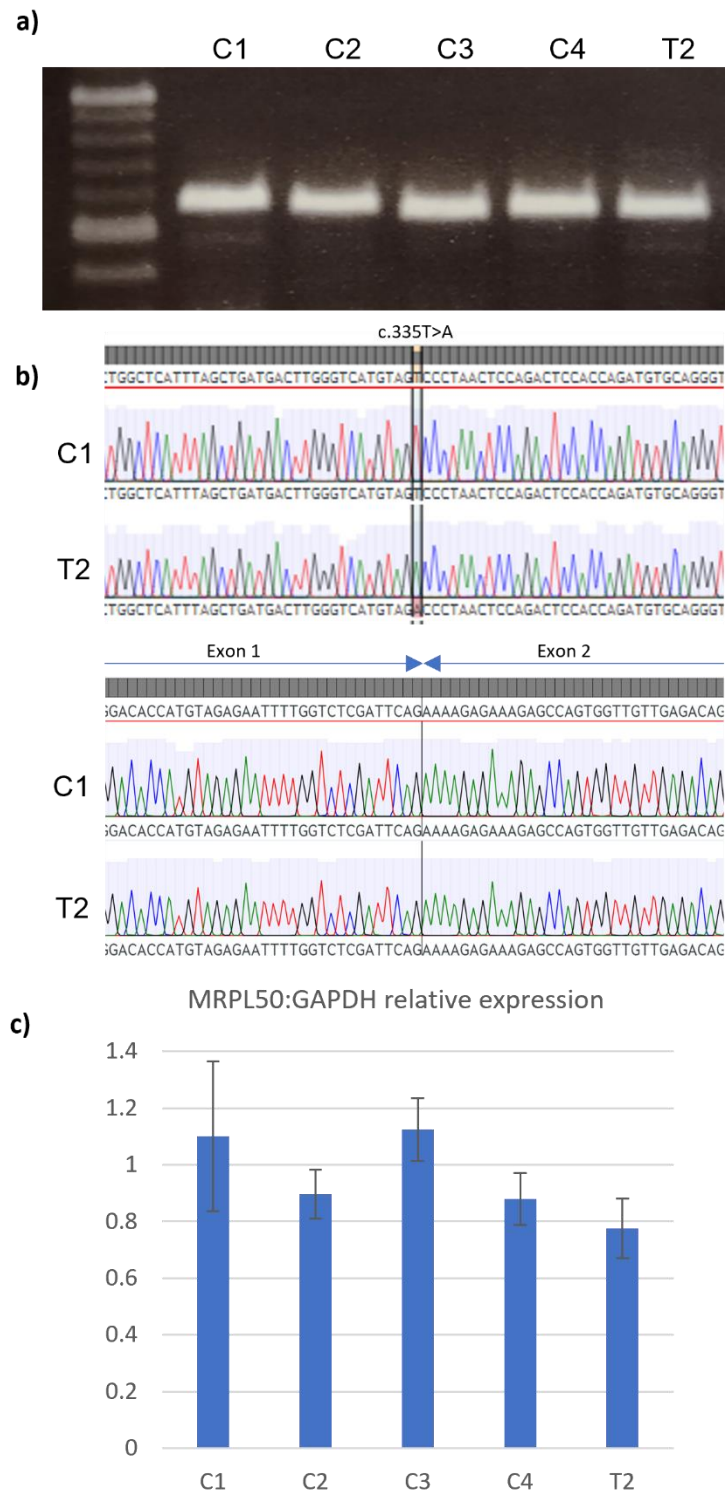
Twin 1



Twin 2



Supplementary Figure S1: Pure-tone audiometry results for Twin 1 (upper) and Twin 2 (lower). The Y-axis (vertical) shows the intensity of the sound, in decibels (dB HL). The X-axis (horizontal) shows the frequency of sound in Hz. Crosses indicate air-conduction thresholds for the left ear, and circles indicate air-conduction thresholds for the right ear. The symbol \triangleright is used to show bone conduction thresholds. An arrow symbol indicates no response to sound at that frequency.



Supplementary Figure S2. The MRPL50 variant does not have a significant impact on MRPL50 transcript expression. a) Full-length RT-PCR of control (C1-C4) and patient (T2) lymphoblast cDNA demonstrates no discernable disruption to MRPL50 splicing b) Chromatograms show the homozygous missense variant in patient cDNA (upper) and normal MRPL50 exon-exon boundary indicating normal splicing (lower). c) qRT-PCR demonstrates that relative MRPL50 expression in T2 does not differ significantly from that in controls (C1-C4). Bars represent the mean of three independent experiments, each performed in triplicate. Error bars: ± 1 SEM.