Destabilization of the IFT-B cilia core complex due to mutations in IFT81 causes a Spectrum of Short-Rib Polydactyly Syndrome

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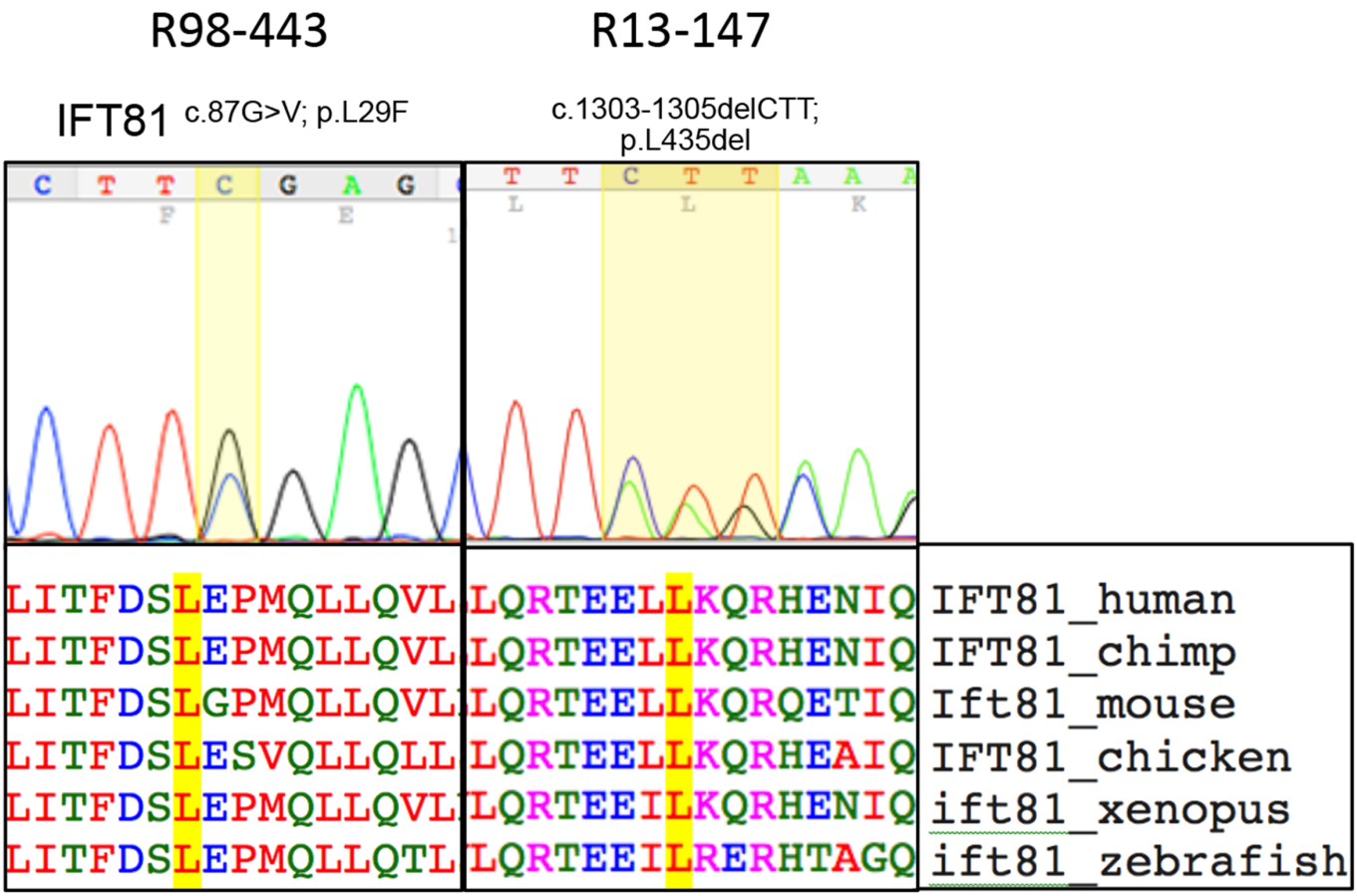
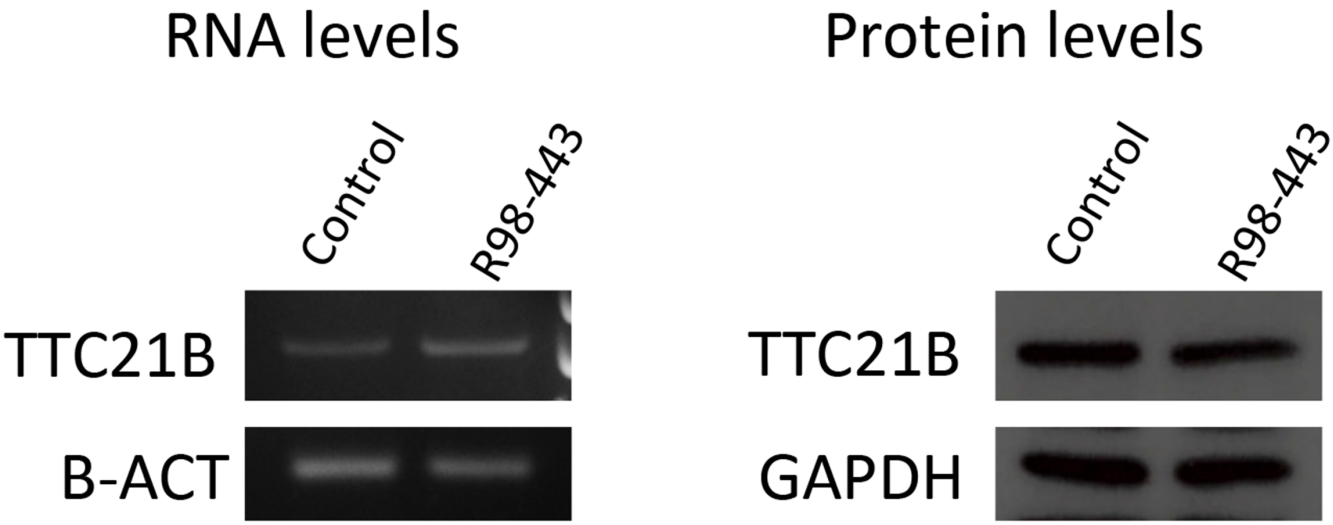
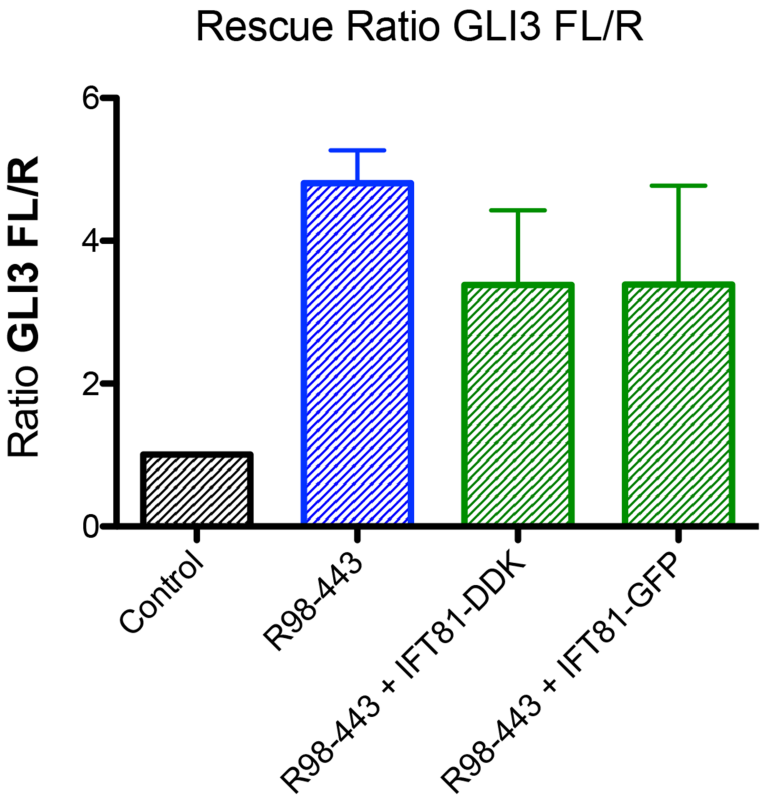


Supplemental Figure 1. Chromatograms demonstrating the missense and deletion mutations in R98-443 and R13-147 highlighting the conservation of the each respective residue across species.



Supplemental Figure 2. RT-PCR and western blot analysis of TTC21B in R98-433 showing that the *TTC21B* variant (c.2600G>A; p.Arg867His; rs76726265) had no apparent effect on cDNA or protein levels in R98-443.



Supplemental Figure 3. Ratio of FL/R forms of GLI3 in R98-443 rescued with expression of wild type IFT81.