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Supplemental Data

A Nonsense Mutation in the Human Homolog

of *Drosophila roghi* Causes Kohlschutter–Tonz Syndrome

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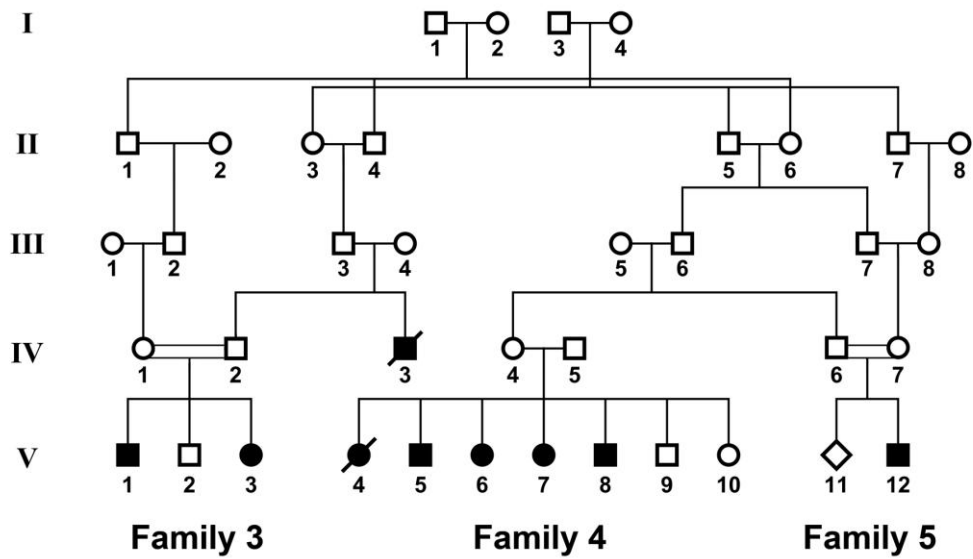
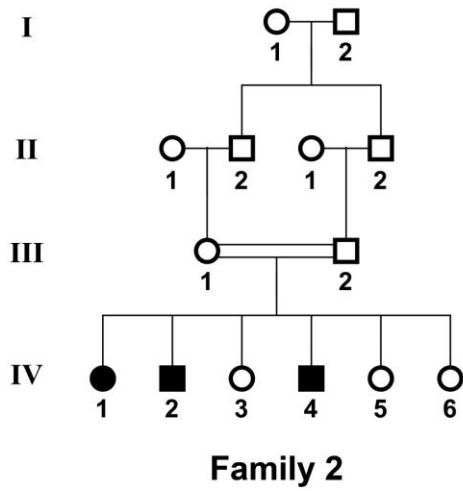
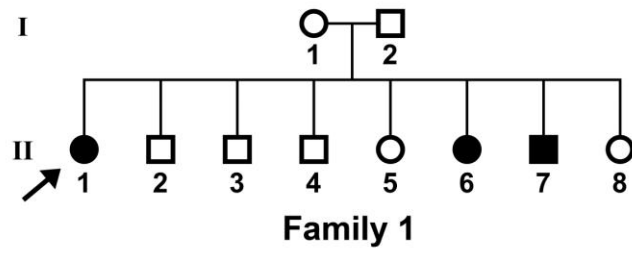


Figure S1. Pedigrees of Participating Families

Three out of the five participating families (families 3-5), relate to one large pedigree with two ancestral origins. Index case, individual 1-II of family 1 is marked with an arrow.

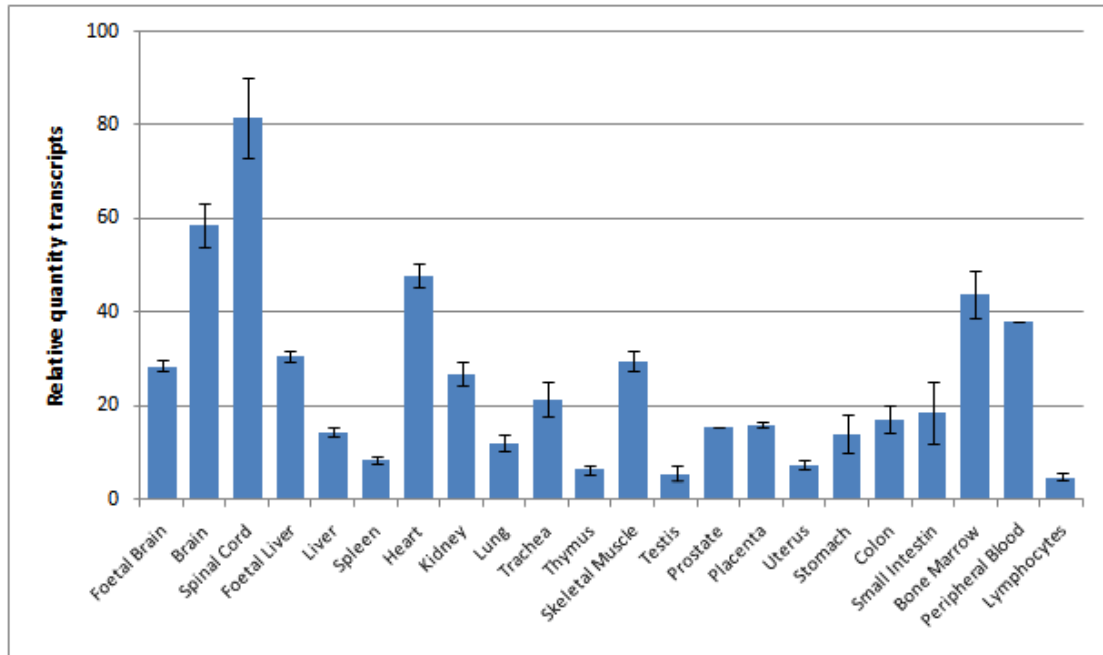


Figure S2. Expression Analysis of the Human *ROGDI* in Different Human Tissues, as Assessed by Means of Quantitative RT-PCR Analysis

ROGDI transcripts are found at a higher level in adult brain, spinal cord, peripheral blood, heart and bone marrow, and at lower but significant levels in other tissues including fetal brain. The ubiquitously expressed *ERCC3* gene (excision repair cross-complementing rodent repair deficiency, complementation group 3) was used as an internal control to normalize the data. Values are the mean \pm SD of three independent experiments. 500 ng of total RNA from different human tissues obtained from Clontech (Takara Bio Europe/Clontech, Saint Germain en Laye, France) and from BioChain Institute (Newark, CA, USA) were primed with 2.5 mM of oligodT and then subjected to reverse transcription with the Reverse Transcriptase kit from Roche, following manufacturer's conditions. cDNAs were amplified in the light Cyler LC480 (Roche / Boehringer, Mannheim, USA) with the LC480 probe master mix (Roche) using a forward primer in exon 10 (5' GACGCCCTGGTCTACTTCAC 3') and a reverse primer in exon 11 (5'TGTGATCAGAAGGGTCTGTAGC 3').