

Supplemental Data

Mutations in *ROGDI* Cause Kohlschütter-Tönz Syndrome

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Table S1. Autozygous Regions in Family A

Chromosome	Flanking SNPs	Size	Number of Protein Coding Genes
3q13.31	rs2134655- rs2866121	4,561,887 bp	33
11q24.1-q24.2	rs763640- rs718520	1,420,060 bp	64
16p13.3	rs2107321- rs1424125	3,255,290 bp	104
17q25.1-q25.3	rs718072-rs2045660	6,592,024 bp	125
Total		15,829,261 bp	326

Table S2. Summary of the Filtering Steps Used to Reduce the Number of Candidate Variants

Description	# of Variants	Genes
total SNPs	20454	
total DIPs	1208	
total Variants	21662	
auto_annovar		
Remove synonymous and non-frameshift variants	9970	
Remove conserved variants from 44-species alignment	4711	
Remove variants in segmental duplication regions	4164	
Remove variants in 1000 genomes Project (2010_11)	913	
Remove variants in dbSNP build 132	791	
Region of interest		
chr13 (113.858.201-118.420.088)	0	
chr11 (123.846.833-125.266.893)	0	
chr16 (2.807.263-6.062.553)	2	<i>ROGDI, CREBBP</i>
chr17 (70.584.327-77.176.351)	5	<i>SLC39A11, ITGB4, EVPL, MGAT5B, USP36</i>
Remove heterozygous variants		
chr16 (2.807.263-6.062.553)	1	<i>ROGDI</i>
chr17 (70.584.327-77.176.351)	1	<i>EVPL</i>

Table S3. Sequences of Primers Used for Genomic PCR Amplification of *ROGDI*

Exons	Primers (including universal M13 sequences in lower case)
1_2 fw.	gtaaaacgacggccagtCAGTCTGGTTCCAGGGCTC
1_2 rv.	caggaaacagctatgacGAGCCAGGGAAATGAGGAG
3_4 fw.	gtaaaacgacggccagtGGCTTCTGGAAGGAGCACTG
3_4 rv.	caggaaacagctatgacGGGAGCTCCCATGCTCTTC
3_4 internal fw.1	AGTGGGCGCTACCAGCTC
3_4 internal fw.2	CAGACCAGCTGGTTGTGC
3_4 internal rv.	GCACAACCAGCTGGTCTG
5 fw.	gtaaaacgacggccagtTAAGGGCTCAGAGTGGCTGT
5 rv.	caggaaacagctatgacCCCAGCTTTGCTGTTAGGA
6 fw.	gtaaaacgacggccagtAGTGTGGAGAGTGGAATGACG
6 rv.	caggaaacagctatgacCCAGGTGTGATTCCGGAT
7 fw.	gtaaaacgacggccagtGAGCTACCATGTCTGTTGTTCC
7 rv.	caggaaacagctatgacCTATAATCTGGGAGTGGCACC
8_9_10 fw.	gtaaaacgacggccagtGCAGACTCCATGAGTGACTGC
8_9_10 rv.	caggaaacagctatgacCTGAGGGTGTGGCAAGGA
11 fw.	gtaaaacgacggccagtGACATATTGTCCTTGGGTGACC
11 rv.	caggaaacagctatgacCTCCTGGCACTTCCAGTGTC

Table S4. Sequences of Primers Used for RT-PCR Analyses of *ROGDI*

	RT-PCR Sequencing	Quantitative RT-PCR
Forward	GAAGCTGCAGCAGATCCAG	CCAAGCAAGAGAACTTCATCC
Reverse	GAGAGCCCCACTCGAACATG	GCATCTTCAGGTTACATCC