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Supplemental Data

Homozygous and Compound Heterozygous Mutations in *TGDS* Cause Catel-Manzke Syndrome

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Table S1. The informative SNVs used for haplotype reconstruction. All positions are with respect to the haploid human reference sequence GRCh37. *TGDS* is on the reverse strand thus all alternative alleles would be the complementary base in *TGDS* transcripts. The position with the pathogenic allele c.298T is coloured in red. A "0/1" represents a heterozygous genotype. A "0/0" means that the individual is homozygous for the reference allele and "1/1" means that the individual is homozygous for the alternative allele. For the haplotype reconstruction a DNA sample of individual 6 was subjected to exome sequencing following the protocol described for individual 1 and 7.

CHR	POS	dbSNP	Ref	Alt	MAF	6-II-2	1-II-1	5-I-3	4-II-4	2-II-2	7-II-1
13	95228658	.	T	C	0	0/0	0/1	0/0	0/0	0/0	0/0
13	95230384	.	A	G	0	0/1	0/0	0/0	0/0	0/0	0/0
13	**95243122**	**rs140430952**	**C**	**A**	**0**	**0/1**	**0/0**	**1/1**	**1/1**	**0/1**	**0/1**
13	95243123	rs61741685	G	A	0.012	0/0	0/1	0/0	0/0	0/0	0/0
13	95243126	.	A	C	0	0/0	0/0	0/0	0/0	0/1	0/0
13	95243148	.	TTC	T	0	0/0	0/1	0/0	0/0	0/0	0/0
13	95243270	rs9301979	G	A	0.321	1/1	1/1	1/1	1/1	0/1	0/1
13	95248348	rs34991132	C	T	0.006	0/0	0/1	0/0	0/0	0/0	0/0
13	95264604	rs9524559	C	T	0.291	0/0	0/0	0/0	0/0	0/1	0/1
13	95275576	rs4296139	C	G	0.433	0/1	1/1	0/0	0/0	0/1	0/0
13	95279505	rs9524568	T	A	0.251	0/0	0/0	0/0	0/0	0/1	0/0

Table S2. The most likely 6 haplotypes H. H1 is the haplotype with the pathogenic allele c.298T.

H1: 0010001 0000 7.000000
H2: 00000000100 1.000000
H3: 00000010010 1.000000
H4: 00001000111 1.000000
H5: 01000010010 1.000000
H6: 10010111010 1.000000

Table S3. The predicted haplotypes for all affected individuals. In agreement with the hypothesis of a founder mutation, all affected individuals with the c.298T allele share the same 50kb Haplotype H1.

1-II-1: (H3, H6)
2-II-2: (H1, H4)
4-II-4: (H1, H1)
5-I-3: (H1, H1)
6-II-2: (H1, H5)
7-II-1: (H1, H2)