Homozygous deletion of Tenascin-R gene in a patient with intellectual disability

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Supplementary Tables.

Table S1. Forward and reverse primers used to amplify *TNR* exons (RefSeq NM_003285.2)

Primer	Exon targeted	Forward	Reverse	Amplicon Size (bp)
TNR.1	1	TGGTTTCCGTTGCAGATTC	GCATTGGTCCTTCTTGTTCAC	663
TNR.2	2	GAGTAGAGAGGCCTTTGCCC	TCCCAAGCTAACTGGAGGC	642
TNR.3	3	TGAGCCCAGCAGGACATATAG	GGGCAATAACTATTCCAGGC	501
TNR.4	4	TGGAGGGAGTTCACCAAGAC	CCCCTGATTCCAAGCTAAAG	277
TNR.5	5	GTCAGCATGGAGGAAAGAGAC	ACTCCTTGGGCAATTCTGTG	368
TNR.6	6	GAAAAGAGAGAGCTGACACCC	CCTCATTCCTAATACTCCTCTTGC	438
TNR.7	7	TCAGAGTTCAAGATGCACCAC	GGCCATGGGTTTGTAAGATG	354
TNR.8	8	TTTTCTTCCCACTTTCAGCC	GGACGAGCCTCCATGACTC	235
TNR.9	9	TTGGAAACACACAAAGCAGG	TGCCTGAATGAATTTCTCACAC	417
TNR.10	10	GAGAGCAGGTCCCTTTAGCC	GAAATCCCCTACTCCTTGGG	467
TNR.11	11	TTGGTCAACATAAAACCAGGG	TTCTGCGGGATATGCTAGTTC	281
TNR.12	12	TAGACTGTACCATGGGCTGC	ACAAGCCCGAACAACTTCAC	288
TNR.13	13	AACTTCTGAGATCATTCCCTGC	GTTTATTCACATCATACTGGTCTGC	275
TNR.14	14	TGTGAAATACCTTTGGATTTGG	AAGTGATAATCTAGTCCACCACAAG	313
TNR.15	15	CCCATACCTGAAATCAAGCAG	CCCTCCAATGTCCTGTGG	294
TNR.16	16	ATGGAGAGGATGACAATGGC	AAAAGAAGGAGGAGGCAACTG	391
TNR.17	17	ACCAGGAGGCCTGCTAAGAC	AGGATGGTGAAGTTGGCTTG	338
TNR.18	18	CAGTTTATTTTCCTTAGCATCTCTTTG	AGCTTCTCAACAAACACAGGG	285
TNR.19	19	CACAGACGCATTTCCCTCC	GGTGCTGCTGCTTCCTGAG	332
TNR.20	20	TTCCATAAGTGGAAGCCAGG	TTTAAAGGGAAATGAAGGACTCAC	375
TNR.21	21	CATTGTGGATTCCCTGTTCC	CACATTGCTATTACCCTCCCC	305

Table S2. TNR variants identified from the 188 ID patients screened.

Hg19 Chrom Position	state	dbSNP131 rs#	Frequency (n=188)	cDNA change	Amino Acid Change	SIFT	PolyPhen	Transmission*
Chr1:175375802	homo+hetero	859398	145	c.49A>G	I17V	1.00	0.13	not tested
Chr1:175375779	homo+hetero	NA	2	c.72C>T	S24S	NA	NA	not tested
Chr1:175375683	homo+hetero	859399	144	c.168A>G	T56T	NA	NA	not tested
Chr1:175375635	homo+hetero	2239818	22	c.210C>T	N72N	NA	NA	not tested
Chr1:175375513	hetero	NA	1	c.338C>T	T113I	NA	0.08	healthy Father
Chr1:175375469	homo+hetero	2239819	74	c.382G>T	A128S	0.71	0.28	not tested
Chr1:175375434	hetero	NA	1	c.417C>T	S139S	NA	NA	not tested
Chr1:175372714	hetero	61731112	1	c.538A>C	N180H	0.16	1.39	not tested
Chr1:175372651	hetero	NA	1	c.601G>A	E201K	0.24	1.26	healthy mother
Chr1:175363013	hetero	NA	1	c.1259G>A	G420E	0.09	0.37	healthy mother
Chr1:175355391	homo+hetero	859437	103	c.1544T>C	T518T	NA	NA	not tested
Chr1:175348815	hetero	NA	1	c.1836C>T	L612L	NA	NA	not tested
Chr1:175348723	homo+hetero	859427	101	c.1928G>A	R643K	0.27	0.07	not tested
Chr1:175335234	homo+hetero	1385541	82	c.2094G>A	S698S	NA	NA	not tested
Chr1:175335162	homo+hetero	7516376	7	c.2166A>G	P722P	NA	NA	not tested
Chr1:175331822	hetero	61731107	2	c.2831G>C	R944P	0.87	0.58	not tested
Chr1:175324717	hetero	NA	3	c.3171C>T	A1057A	NA	NA	not tested
Chr1:175324711	homo+hetero	61731115	38	c.3177C>T	I1059I	NA	NA	not tested
Chr1:175324651	homo+hetero	2228359	94	c.3237T>C	D1079D	NA	NA	not tested
Chr1:175323594	hetero	35460270	1	c.3456C>T	Y1105Y	NA	NA	not tested
Chr1:175306758	hetero	61731114	2	c.3440C>T	T1147I	0.07	1.46	not tested
Chr1:175306724	hetero	NA	1	c.3474G>A	G1158G	NA	NA	not tested
Chr1:175299301	homo+hetero	2027867	75	c.3702A>G	Q1234Q	NA	NA	not tested
Chr1:175299249	hetero	35421365	1	c.3754C>T	L1252L	NA	NA	not tested
Chr1:175293570	homo+hetero	NA	2	c.3879G>A	S1293S	NA	NA	not tested

*Transmission was tested only on unique variants (frequency = 1). NA, not available. SIFT scores (http://sift.jcvi.org): damaging <0.05, tolerated >0.05. PolyPhen scores (http://genetics.bwh.harvard.edu/pph/): damaging > 1.5, tolerated < 1.5