

Supplemental Table 1a. Expression of genes located in 15q13.3 assessed by microarray

Transcript Cluster ID	Gene Symbol	RefSeq	Proband (double deletion)			Unaffected brother			Control boys (n=3)		
			p-value	Fold-Change	Raw data	p-value	Fold-Change	Raw data	Raw data	Raw data	Raw data
3586834	MTMR15	NM_014967	0.07	-1.55	31.10	0.81	1.03	88.79	96.11	70.71	87.24
3587015	KLF13	NM_015995	0.03	-1.74	130.89	0.32	1.13	780.57	722.93	575.60	693.16
3587226	CHRNA7	NM_000746	0.61	-1.04	121.88	0.18	1.15	156.68	116.76	137.54	134.98
3615985	MTMR10	NM_017762	0.00	-1.75	44.12	0.19	-1.05	215.50	242.14	227.50	229.73
3616044	TRPM1	NM_002420	0.45	1.05	40.50	0.13	1.15	45.21	39.31	39.09	34.59
3616216	OTUD7A	NM_130901	0.76	-1.02	109.47	0.73	1.02	115.82	105.64	120.16	111.64

Mother			Grandmother			Control females (n=3)		
p-value	Fold-Change	Raw data	p-value	Fold-Change	Raw data	Raw data	Raw data	Raw data
0.25	-1.53	54.79	0.31	-1.44	67.15	86.66	165.16	169.28
0.22	-1.32	517.21	0.26	-1.27	562.56	689.39	873.70	1029.51
0.47	-1.07	150.23	0.45	-1.07	149.38	154.71	182.39	158.87
0.31	-1.20	136.79	0.17	-1.33	109.51	218.16	154.11	186.12
0.77	1.02	37.96	0.50	1.05	39.45	38.92	37.99	33.85
0.32	-1.09	108.32	0.57	-1.05	115.73	124.45	113.12	133.28

Father			Uncle			Control males (n=3)		
p-value	Fold-Change	Raw data	p-value	Fold-Change	Raw data	Raw data	Raw data	Raw data
0.22	-1.48	50.81	0.65	-1.13	97.50	79.77	139.48	133.44
0.10	-1.37	475.49	0.03	-1.76	159.98	738.71	951.06	922.26
0.56	-1.05	141.88	0.05	1.35	219.99	137.89	155.68	165.84
0.62	-1.18	140.61	0.50	-1.27	121.51	260.15	133.90	160.84
0.69	-1.02	42.62	0.01	1.61	74.06	42.48	46.36	42.58
0.37	-1.22	95.68	0.54	1.14	160.36	101.20	158.82	145.23

Supplemental Table 1b. Expression of TRPM1 and CHRNA7 assessed by QRT-PCR

	TRPM1		CHRNA7	
	C _T	fold change	C _T	fold change
Proband	undetectable		undetectable	
Control boys	35.40		37.91	
Mother	37.30	-5.60 ¹	38.68	-2.30 ¹
Control females	34.51		37.53	
Father	36.60	-4.56 ²	39.49	-4.08 ²
Control males	34.32		37.45	
Brain RNA	33.08	+3.33 ³	32.39	+10.48 ³

- 1 fold change relative to comparison females
- 2 fold change relative to comparison males
- 3 fold change relative to average of all controls