

**The American Journal of Human Genetics, Volume 89
Supplemental Data**

**Assessment of 2q23.1 Microdeletion Syndrome Implicates
MBD5 as a Single Causal Locus of Intellectual
Disability, Epilepsy, and Autism Spectrum Disorder**

Michael E. Talkowski, Sureni V. Mullegama, Jill A. Rosenfeld, Bregje WM van Bon, Yiping Shen, Elena A. Repnikova, Julie Gastier-Foster, Devon Lamb Thrush, Sekar Kathiresan, Douglas M. Ruderfer, Colby Chiang, Carrie Hanscom, Carl Ernst, Amelia M. Lindgren, Cynthia C. Morton, Yu An, Caroline Astbury, Louise A. Brueton, Klaske D. Lichtenbelt, Lesley C. Ades, Marco Fichera, Corrado Romano, Jeffrey W. Innis, Charles A Williams, Dennis Bartholomew, Margot I. Van Allen, Aditi Parikh, Lilei Zhang, Bai-Lin Wu, Robert E. Pyatt, Stuart Schwartz, Lisa G. Shaffer, Bert BA de Vries, James F. Gusella, and Sarah H. Elsea

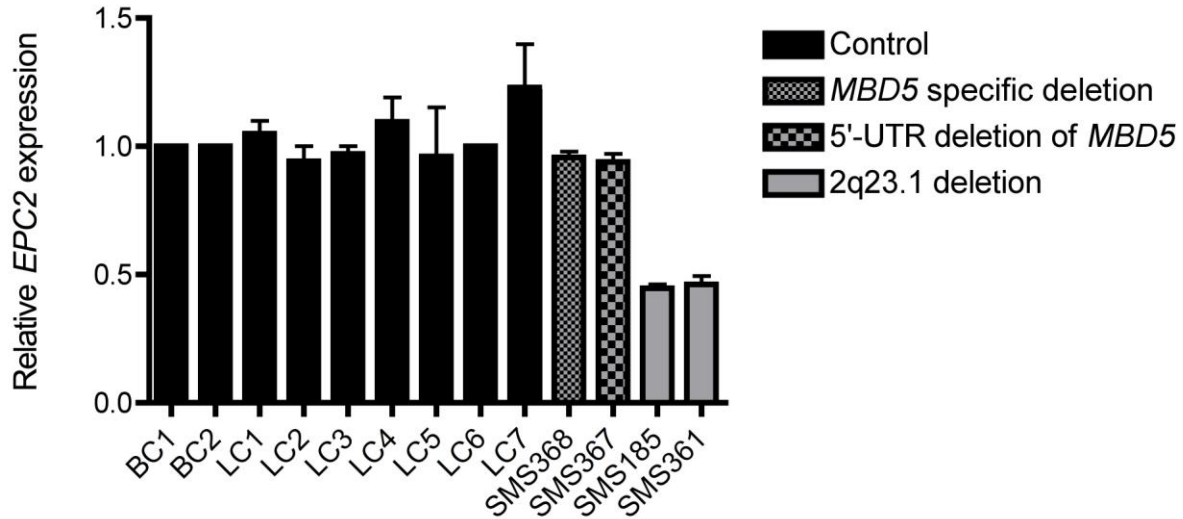


Figure S1. *EPC2* Expression Is Normal in *MBD5*-Specific Deletion Cases

Quantitative real-time PCR (q-RT-PCR) mRNA expression analysis of *EPC2* in lymphoblastoid cell lines or fresh white blood cells from 2q23.1 deletion syndrome individuals with no deletions for *EPC2* but having *MBD5* deletions was done (SMS367, SMS368). Fresh white blood cells or lymphoblastoid cell lines from nine unaffected (no *MBD5* deletion) subjects were used as normal controls. Results were normalized to *GAPDH* expression. Relative expression values are based on the $\Delta\Delta C_t$ value. Expression of all controls was normalized to 1. Each bar represents mean (\pm SEM) of values from 3 independent experiments. Data show a normal range, 0.88 to 1.19-fold *EPC2* expression in lymphocytes (BC1-2) and lymphoblastoid cell lines, (LCL1-4). SMS367 and SMS368 cases were not deleted for *EPC2*, and data show 92-100% expression of *EPC2*, which is a normal range of expression for *EPC2*. SMS185 and SMS361 were previously reported cases with larger 2q23.1 deletions including both *EPC2* and *MBD5* (Williams et al 2010).

Table S1. MBD5 Primer Sequences

Exon	Exon_Start	Exon_Stop	F Primer	Primer Sequence (Chr2+)	F_chr2_strand+	R Primer	Primer Sequence (Chr2-)	R_chr2_strand-
MBD5-001 ex1	148495050	148495085						
MBD5-001 ex2	148495687	148495723	MBD5_1-2F	GCTACAATGTCTTCCGGGTC	148494966 148494985	MBD5_1-2R	GTGGTTTGGTGCTACTGTTGC	148495781 148495801
MBD5-001 ex3	148652739	148652832	MBD5_3F	GTCCTATAATACATCCCTTTGCC	148652603 148652625	MBD5_3R	GGTAACTTAGACAGGAGCAGTG	148652926 148652947
MBD5-001 ex4	148707284	148707434	MBD5_4F	CAAGCCCTTTCTGTTAGAGTC	148707134 148707154	MBD5_4R	CATGCATACATAAATTCCTACAGG	148707535 148707558
MBD5-001 ex5	148816253	148816375	MBD5_5F	CTGAAGGATGAAGATACTAATACC	148816100 148816123	MBD5_5R	CAATCTATAGATGTCAGACACTTCC	148816455 148816479
MBD5-001 ex6	148932242	148932910	MBD5_6F	TGTGATTGGGTACACTTCGTAG	148932141 148932162	MBD5_6R	TGCCATCAGTCACCATGCTTG	148932973 148932993
MBD5-001 ex7	148936621	148936723	MBD5_7F	ATATCCCATAAAGGACAGTAAGAC	148936521 148936544	MBD5_7R	GATACAGCTCTAATGAAAAGGTAG	148936969 148936992
MBD5-001 ex8	148937778	148937958	MBD5_8F	CTGTTAGAACATCAGGAGGTATC	148937480 148937502	MBD5_8R	GCATTGTGCGTGCCTGAAAAG	148938048 148938068
MBD5-001 ex9	148942380	148944500	MBD5_9F	CACCACAAAAGAGTTGAGACTG	148942325 148942346	MBD5_9R	TCCCAATAGACAAATCCCTAGC	148944656 148944677
MBD5-001 ex10	148957149	148957475	MBD5_10F	GTTACAATCAATGAAGGTGCCATG	148957022 148957045	MBD5_10R	GCTAAAGCAGCGTTTACATTCC	148957518 148957539
MBD5-001 ex11	148959781	148959989	MBD5_11F	TCGGGTACAAAGAGAGGCATC	148959709 148959729	MBD5_11R	GTGAAAGGGCACGTAACCTAC	148960074 148960094
MBD5-001 ex12	148963425	148964633	MBD5_12F	CTGAGTACGTTTTCTGTCCAGC	148963125 148963146	MBD5_12R	TCCAGAAGCTTGTGAGACCTC	148964752 148964772
MBD5-001 ex13	148976475	148976548	MBD5_13F	GGACAGAAGATTTGACAAGGTAG	148976332 148976354	MBD5_13R	GCTTGTCTATAATGGTCCTCTTTG	148976677 148976700
MBD5-001 ex14	148984099	148984174	MBD5_14F	GGCTTTCTCTCGTGGAATTGG	148983994 148984014	MBD5_14R	CAGAAAGCTGTGGTTTGAAC TTC	148984438 148984460
MBD5-001 ex15	148986909	148987490	MBD5_15F	GCATTCTGCATTCTCTGTAGTG	148986710 148986731	MBD5_15R	CAGTGGTGTGCAGTGTATGTC	148987576 148987596

Bold = non-coding exon.