

Table S1: ATR-X methylation: Significant regions detected by methylation array in ATR-X patients (n=17) compared with controls (n=210) using cut-off of 0.15 methylation difference

Chr	# Probes	Estimate	F value	p-value	Within CpG island	Nearest gene	Distance to nearest gene (bp)
chr1	3	0.178	118.3	0.000045	No	<i>CCDC181</i>	0
chr1	5	0.228	107.7	0.000045	No	<i>C1orf35</i>	453
chr2	4	-0.186	66.9	0.000045	Yes	<i>GGT8P</i>	30163
chr2	3	-0.154	54.0	0.000045	No	<i>TTN-AS1</i>	0
chr2	3	0.162	94.7	0.000045	Yes	<i>BOK-AS1</i>	3287
chr3	6	0.271	405.3	0.000045	Yes	<i>CD47</i>	415
chr3	4	0.271	187.5	0.000045	No	<i>DPPA4</i>	0
chr4	5	0.217	65.7	0.000045	Yes	<i>ZNF718</i>	0
chr5	13	-0.225	89.6	0.000045	No	<i>PRDM9</i>	0
chr5	7	0.197	106.0	0.000045	Yes	<i>BHMT2</i>	0
chr5	5	0.231	581.8	0.000045	No	<i>CAMLG</i>	581
chr5	9	0.335	303.4	0.000045	Yes	<i>ZNF300</i>	0
chr5	3	0.197	184.5	0.000045	Yes	<i>ADAM19</i>	0
chr6	6	-0.179	63.6	0.000045	No	<i>QKI</i>	507222
chr6	5	-0.295	145.1	0.000045	No	<i>LOC401286</i>	21624
chr7	3	-0.271	105.5	0.000045	No	<i>GPR85</i>	0
chr8	3	0.236	203.7	0.000045	No	<i>POTEA</i>	11130
chr10	4	0.246	149.3	0.000045	Yes	<i>ZNF248</i>	0
chr10	3	-0.300	232.2	0.000045	No	<i>RASGEF1A</i>	0
chr10	3	-0.189	60.0	0.000045	No	<i>INPP5A</i>	18901
chr11	11	0.153	82.3	0.000045	Yes	<i>MIR4492</i>	0
chr12	6	0.163	165.7	0.000045	Yes	<i>ALG10B</i>	177834
chr13	3	-0.196	61.2	0.000045	No	<i>ATP11AUN</i>	26813
chr16	3	-0.176	72.9	0.000045	Yes	<i>LINC00273</i>	2269
chr18	5	0.198	98.1	0.000045	Yes	<i>LOC100131655</i>	7094
chr19	6	0.175	210.8	0.000045	Yes	<i>PPAP2C</i>	0
chr19	4	0.156	74.8	0.000045	Yes	<i>MOB3A</i>	0
chr19	3	0.260	137.8	0.000045	Yes	<i>C3</i>	0
chr19	5	0.157	64.8	0.000045	Yes	<i>RGL3</i>	0
chr19	7	0.222	221.6	0.000045	Yes	<i>LOC100289333</i>	0
chr19	8	0.198	385.7	0.000045	No	<i>ZNF486</i>	0
chr19	8	0.159	63.8	0.000045	Yes	<i>ZNF274</i>	0
chr20	4	0.266	154.5	0.000045	Yes	<i>SRXN1</i>	3837
chr21	3	0.288	64.7	0.000045	Yes	<i>SPATC1L</i>	0
chrY	4	0.271	61.5	0.000045	Yes	<i>BCORP1</i>	0
chr11	7	0.172	83.3	0.000089	Yes	<i>LOC441601</i>	0

chr10	4	-0.185	80.7	0.000134	No	<i>STK32C</i>	0
chr8	9	0.273	124.2	0.000223	Yes	<i>POTEA</i>	15068
chr6	5	-0.205	91.6	0.000268	Yes	<i>PACSIN1</i>	0
chr6	6	0.192	51.6	0.000357	Yes	<i>KIF25</i>	0
chr1	4	0.155	67.0	0.000669	Yes	<i>TFB2M</i>	0
chr16	6	0.212	406.6	0.001115	Yes	<i>ATF7IP2</i>	0
chr5	13	0.233	237.0	0.002274	Yes	<i>ZNF300P1</i>	0
chr1	10	-0.151	115.9	0.002720	No	<i>UBE2U</i>	0
chr18	5	-0.163	57.4	0.002720	Yes	<i>CTDP1</i>	62202

Significant regions: Probes>3, Estimate > 15%, F value>50, *p*-value<0.01.