

Additional file 2

Kabuki syndrome type 1 and type 2 converge on a shared chromatin and transcriptional state in hiPSC-derived neuroectoderm

Figure S6. Per-read evidence at the edited positions.

Every read overlapping each engineered position, taken from the RNA-seq and ATAC-seq alignments and classified by the sequence spanning the edit: wild-type allele ("SANE"), edited allele ("MISMATCH" relative to the reference) or reads that do not cover the edited base ("NOT_COVERING"). The edited base is highlighted and the reference sequence is shown along the top of each panel; the header of each panel gives the sample, the assay and the window. **(A)** The *KMT2D* locus, chr12:49,052,371-49,052,391, for the wild type and the three *KMT2D*_Het clones (pages 2-13). **(B)** The *KDM6A* locus, chrX:44,961,342-44,961,386, for the wild type and the three *KDM6A*_ko clones (pages 14-19). Counts per class are given in each panel header. Coverage at the *KDM6A* position is low in every sample: the wild-type samples carry 4, 6 and 7 wild-type reads, *KDM6A*_ko_1 carries three reads, all edited, and no read spans the edited base in *KDM6A*_ko_2 or *KDM6A*_ko_3.

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 1 of 12

WT_1 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	reference
SANE (n=28)																				
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	79:12066:18566 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	295:13897:2457 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	295:13897:2457 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	58:47169:18374 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	79:12066:18566 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	296:26553:2121 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	60:30991:13106 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	60:30991:13106 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	58:46985:24955 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	294:6741:14114 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	163:19157:1512 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	87:37564:21160 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	40:13351:23386 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	2272:3099:5468 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	56:31619:14451 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	40:13351:23386 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	56:31619:14451 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	47:33311:27629 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	47:33311:27629 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	87:37564:21160 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	T	G	G	G	G	C	C	T	38:41891:11424 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	2272:3099:5468 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	62:23419:28910 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	80:27228:23402 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	80:27228:23402 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	81:34300:18838 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	17:13786:23658 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	T	G	G	G	G	C	C	T	17:13786:23658 win=TCCTCGTTTA
MISMATCH (n=2)																				
T	C	T	C	C	T	C	G	-	-	-	-	-	-	-	-	-	-	-	-	14:46707:11985 win=TCCTCGA
T	C	T	C	C	T	C	G	-	-	-	-	-	-	-	-	-	-	-	-	14:46707:11985 win=TCCTCGA
NOT_COVERING (n=24)																				
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	130:5946:24683
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	130:5946:24683
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	51:30667:18854
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	176:4393:18662
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	194:48908:5067
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	81:13675:11937
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	2252:6556:9887
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	176:4393:18662
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	2252:6556:9887
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	201:7037:22281
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	77:10827:24971
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	77:10827:24971
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	93:27792:17525
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	238:49502:5468
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	51:28310:17910
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	28:44562:16180
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	51:28310:17910
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	28:44562:16180
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	101:6741:11969
-	-	-	C	C	T	C	G	T	T	T	A	G	G	G	G	G	C	C	T	82:17206:23274
-	-	-	-	-	-	-	-	-	T	T	A	G	G	G	G	G	C	C	T	294:6741:14114
-	-	-	-	-	-	-	-	-	-	-	-	G	G	G	G	G	C	C	T	62:23419:28910
-	-	-	-	-	-	-	-	-	-	-	-	-	G	G	G	G	C	C	T	58:46985:24955
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	T	150:17179:1224

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 2 of 12

WT_1 ATAC -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

	T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=2)																						
	T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	102:5337:17529 win=TCCTCGTTTA
	T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	102:20709:6680 win=TCCTCGTTTA
NOT_COVERING (n=1)																						
	.	.	.	.	.	.	.	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1501:4784:4909

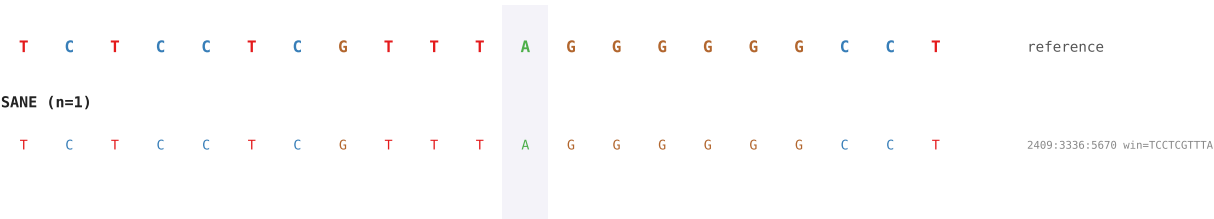
(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 3 of 12

WT_2 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=27)																					
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	23:48177:12097 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	295:43102:4939 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	12:39533:18934 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	47:42593:22922 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	24:39755:24571 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	132:8488:25179 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	198:39062:2489 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	250:29382:1192 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	181:2775:29695 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	209:8026:12978 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	52:23493:18662 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	37:48159:12161 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	22:20904:27950 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	37:48159:12161 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	22:20904:27950 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	52:23493:18662 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	79:12982:20279 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	141:39376:7934 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	155:2304:20616 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	221:47003:5644 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	221:47003:5644 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	92:29780:12769 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	04:13425:12978 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	04:13425:12978 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	155:2304:20616 win=TCCTCGTTTA
-	-	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	237:8766:22970 win=TCCTCGTTTA
-	-	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	237:8766:22970 win=TCCTCGTTTA
MISMATCH (n=3)																					
T	C	T	C	C	T	C	G	-	-	-	-	-	-	-	-	-	-	-	-	-	110:52051:6604 win=TCCTCGA
T	C	T	C	C	T	C	G	G	T	T	A	G	G	G	G	G	G	C	C	T	141:39376:7934 win=TCCTCGG
T	C	T	A	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	92:29780:12769 win=TACTCGTTTA
NOT_COVERING (n=14)																					
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	298:2572:23898
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	14:14100:25420
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	90:38322:26957
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	90:38322:26957
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	07:49573:16500
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	74:51404:12529
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	74:51404:12529
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	66:39496:19094
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	66:39496:19094
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	1156:1398:8574
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	1194:8193:5035
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	87:35567:22057
T	C	T	C	C	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	294:21052:8350
-	-	-	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	79:12982:20279

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 4 of 12

WT_2 ATAC -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

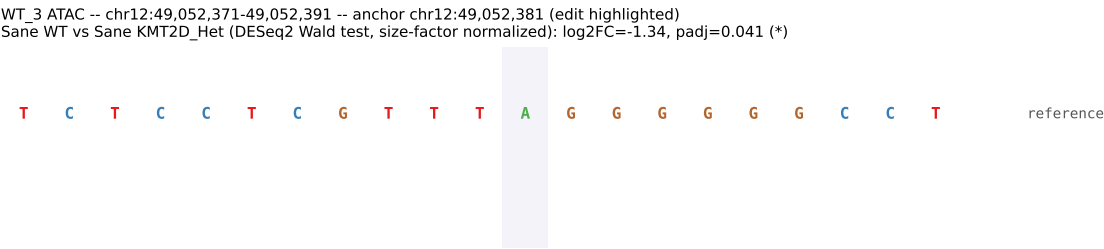


(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 5 of 12

WT_3 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=42)																					
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	218:21237:9951 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	218:21237:9951 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	278:12824:7485 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	278:12824:7485 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	-	-	-	-	-	-	-	25:39450:18278 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	-	-	-	-	-	-	-	25:39450:18278 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	45:31453:26524 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	T	G	G	G	G	C	C	-	233:8433:27421 win=TCCTCGTTT
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	58:17817:29391 win=TCCTGTTT
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	20:36445:26653 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	255:6168:23114 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	255:6168:23114 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	266:22060:6380 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	266:22060:6380 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	79:44941:24523 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	18:25028:26220 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	76:46180:17285 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	-	198:14692:2713 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1202:4032:1256 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	-	198:14692:2713 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	140:15015:6188 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	140:15015:6188 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	A	T	83:26525:12065 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	66:50220:11024 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	03:38202:21624 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	03:38202:21624 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	133:40060:4058 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	133:40060:4058 win=TCCTCGTTT
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1202:4032:1256 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	92:31814:11969 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	92:31814:11969 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	66:50220:11024 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	26:14988:23498 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	26:14988:23498 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	51:20331:25996 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	95:18196:10095 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	51:20331:25996 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	95:18196:10095 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	107:44091:5532 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	61:42279:11200 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	107:44091:5532 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	176:6140:13042 win=TCCTCGTTTA
MISMATCH (n=1)																					
T	C	T	C	C	T	C	G	G	T	T	A	G	G	G	G	G	G	C	C	T	83:26525:12065 win=TCCTCGG
NOT_COVERING (n=14)																					
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	175:2757:28318
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	121:9080:21112
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	121:9080:21112
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	38:45940:22857
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	38:45940:22857
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	36:44322:23802
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	176:16735:6989
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	130:2886:25211
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	192:9015:13058
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	290:7869:11424
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	192:9015:13058
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	290:7869:11424
T	C	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	203:33949:9743
T	C	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	203:33949:9743

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 6 of 12

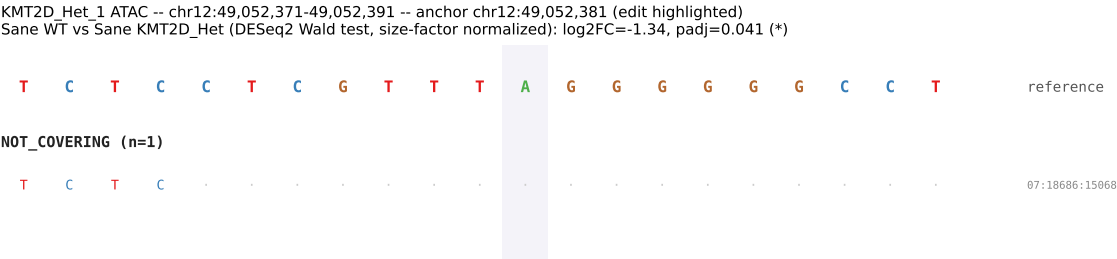


(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 7 of 12

KMT2D_Het_1 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=15)																					
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	-	91:48113:11729 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	-	-	-	-	-	-	-	-	110:9542:17910 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	-	-	-	-	-	-	-	-	110:9542:17910 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	27:37582:21096 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	01:46291:20808 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	01:46291:20808 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	83:48168:13554 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	83:48168:13554 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1226:2387:1705 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	67:44516:25804 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1226:2387:1705 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	67:44516:25804 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	27:37582:21096 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	266:47188:9615 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	266:47188:9615 win=TCCTCGTTTA
MUTATED (n=14)																					
T	C	T	C	C	T	C	G	T	T	T	T	-	-	-	-	-	-	-	-	-	252:5780:11681 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	A	G	G	G	G	G	-	-	271:27542:4507 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	101:4929:11873 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	59:47123:24235 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	271:27542:4507 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	64:43998:15908 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	64:43998:15908 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	59:47123:24235 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	50:18556:26380 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	97:38451:13730 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	97:38451:13730 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	61:45986:29343 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	61:45986:29343 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	G	G	G	G	G	G	C	C	50:18556:26380 win=TCCTCGTTTA
MISMATCH (n=2)																					
T	A	T	C	A	T	C	G	T	T	T	A	G	G	-	-	-	-	-	-	-	80:49962:29503 win=TCATCGTTTA
T	C	G	C	C	T	C	G	T	-	-	-	-	-	-	-	-	-	-	-	-	80:49962:29503 win=GCCTCGTGA
NOT_COVERING (n=17)																					
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	38:45404:10592
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	51:27986:21064
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	245:6436:12529
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	290:45533:4859
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	51:27986:21064
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	245:6436:12529
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	290:45533:4859
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	219:48039:4939
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	219:48039:4939
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	22:40855:19335
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	2265:1666:6092
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	65:38072:28222
T	C	T	C	C	T	C	G	T	T	T	-	-	-	-	-	-	-	-	-	-	91:48113:11729
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	146:28282:8734
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	52:42824:15828
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	146:28282:8734
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	52:42824:15828

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 8 of 12

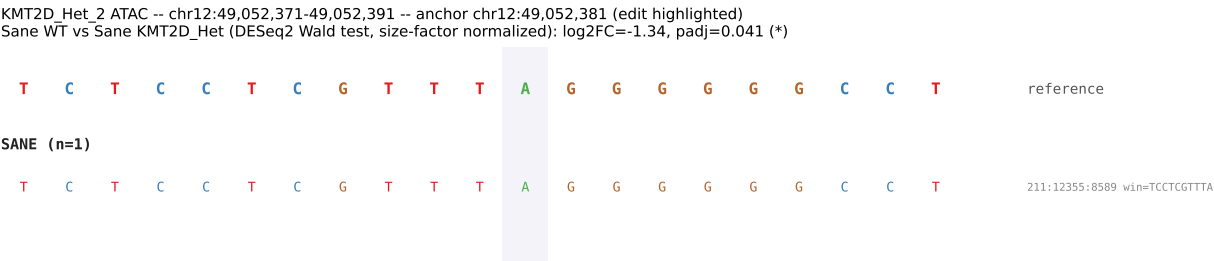


(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 9 of 12

KMT2D_Het_2 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=14)																					
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	-	90:40171:17733 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	-	90:40171:17733 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	262:44359:8654 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	144:4051:22489 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	59:42492:18390 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1112:6436:4875 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	05:30760:28750 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	1112:6436:4875 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	05:30760:28750 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	188:37730:3706 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	43:24131:13010 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	226:45293:7133 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	226:45293:7133 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	43:24131:13010 win=TCCTCGTTTA
MUTATED (n=19)																					
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	164:4837:22345 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	164:4837:22345 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	-	-	-	-	-	-	-	-	-	175:20627:8190 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	21:43425:18406 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	175:20627:8190 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	15:16911:19879 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	32:36270:21576 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	69:18917:24827 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	69:18917:24827 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	T	A	G	G	G	G	-	-	-	270:24066:6012 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	89:20886:22249 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	168:5364:17109 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	32:36270:21576 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	89:20886:22249 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	71:40069:22777 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	24:35280:17845 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	24:35280:17845 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	71:40069:22777 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	55:10393:29695 win=TCCTCGTTTA
MISMATCH (n=2)																					
T	C	T	C	A	T	C	G	T	T	T	A	G	G	G	G	G	G	C	A	T	59:42492:18390 win=TCATCGTTTA
T	C	T	A	C	T	C	G	T	T	T	T	A	G	G	G	G	G	-	-	-	270:24066:6012 win=TACTCGTTTT
NOT_COVERING (n=19)																					
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	197:13120:7709
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	63:13925:16725
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	2297:7277:7261
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	26:15330:10095
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	47:43730:10512
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	47:43730:10512
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	1180:5234:7405
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	48:22707:18742
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	52:45681:24875
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	39:18612:21192
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	264:10790:7709
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	62:30797:14339
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	21:22051:10335
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	21:22051:10335
T	C	T	C	C	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	29:51552:27293
-	-	-	-	-	-	-	G	T	T	T	A	G	G	G	G	G	G	C	C	T	188:37730:3706
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	91:28587:25596
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	55:10393:29695
-	-	-	-	-	-	-	-	T	T	T	A	G	G	G	G	G	G	C	C	T	168:5364:17109

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 10 of 12

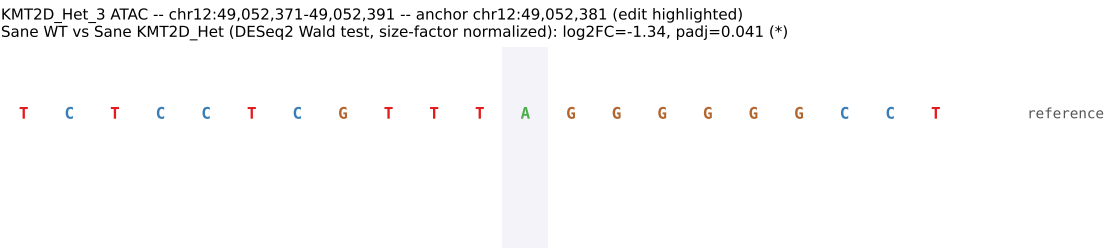


(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 11 of 12

KMT2D_Het_3 RNA -- chr12:49,052,371-49,052,391 -- anchor chr12:49,052,381 (edit highlighted)
Sane WT vs Sane KMT2D_Het (DESeq2 Wald test, size-factor normalized): log2FC=-1.34, padj=0.041 (*)

T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	reference
SANE (n=12)																					
T	C	T	C	C	T	C	G	T	T	T	A	G	G	A	G	G	-	-	-	-	86:11438:15075 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	-	C	C	-	43:29114:18054 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	-	-	-	-	-	-	-	-	-	77:50165:28478 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	195:26350:9423 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	195:26350:9423 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	249:17733:4747 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	249:17733:4747 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	191:11965:1256 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	91:45894:21336 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	91:45894:21336 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	-	-	-	-	-	-	77:50165:28478 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	63:26969:29423 win=TCCTCGTTTA
MUTATED (n=16)																					
T	C	T	C	C	T	C	G	T	T	T	T	-	-	-	-	-	-	-	-	-	294:40569:7982 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	24:47280:21464 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	180:20488:9455 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	180:20488:9455 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	150:24723:2922 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	20:46097:12721 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	121:33533:5628 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	121:33533:5628 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	42:31370:28398 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	42:31370:28398 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	20:46097:12721 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	207:29530:2665 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	207:29530:2665 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	151:35881:3034 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	151:35881:3034 win=TCCTCGTTTA
T	C	T	C	C	T	C	G	T	T	T	T	G	G	G	G	G	G	C	C	T	195:27700:6188 win=TCCTCGTTTA
MISMATCH (n=2)																					
T	C	T	A	C	T	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	195:27700:6188 win=TACTCGTTTT
T	C	T	A	C	T	A	G	T	T	T	A	G	G	G	G	G	G	C	C	T	63:26969:29423 win=TACTAGTTTA
NOT_COVERING (n=27)																					
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	85:41234:21721
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	85:41234:21721
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	29:13508:23114
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	29:13508:23114
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	39:50849:23610
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	39:50849:23610
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	211:5216:11024
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	15:38821:27053
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	98:10116:18870
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	211:5216:11024
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	15:38821:27053
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	98:10116:18870
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	72:45829:22569
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	29:20396:16981
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	98:11521:17813
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	41:38119:10592
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	56:43730:20568
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	11:17992:13938
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	11:17992:13938
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	144:35484:3242
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	144:35484:3242
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	27:45533:22185
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	222:39302:6653
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	222:39302:6653
T	C	T	C	C	T	C	-	-	-	-	-	-	-	-	-	-	-	-	-	-	68:16421:20375
-	-	-	-	-	-	C	G	T	T	T	A	G	G	G	G	G	G	C	C	T	191:11965:1256
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	C	C	T	24:47280:21464

(A) KMT2D locus, chr12:49,052,371-49,052,391 — sheet 12 of 12



(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 1 of 6

WT_1 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

[illegible]

WT_1 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C reference

(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 2 of 6

WT_2 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

SANE (n=6)																														reference															
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	115:49962:4939 win=TAGGTCACCTTCAACCTCTTATTGG
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	22:22661:27853 win=TAGGTCACCTTCAACCTCTTATTGG
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	2231:9746:7757 win=TAGGTCACCTTCAACCTCTTATTGG
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	61:20285:13986 win=TAGGTCACCTTCAACCTCTTATTGG
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	141:44313:1336 win=TAGGTCACCTTCAACCTCTTATTGG
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	141:44313:1336 win=TAGGTCACCTTCAACCTCTTATTGG
NOT_COVERING (n=13)																																													
T	C	T																												12:39385:17301															
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G															261:24982:3626															
															C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	115:49962:4939		
															C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	261:24982:3626		
															C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	297:14498:1929		
															A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	2231:9746:7757			
															C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	119:30122:8398											
															C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	119:30122:8398											
															T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	124:9431:23098												
															T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	124:9431:23098												
															T	G	G	A	A	G	A	T	T	A	T	C	258:16374:9279																		
															T	G	G	A	A	G	A	T	T	A	T	C	258:16374:9279																		
															G	A	A	G	A	T	T	A	T	C	22:22661:27853																				

WT_2 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C reference

(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 3 of 6

WT_3 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	reference
SANE (n=7)	
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	246:24510:4187 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	73:34938:12257 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	139:7453:11825 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	246:24510:4187 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	73:34938:12257 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	70:20276:19207 win=TAGGTCACCTCAACCTCTTATTGG
T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	97:24427:20792 win=TAGGTCACCTCAACCTCTTATTGG
NOT_COVERING (n=11)	
- - - - -	01:50581:11392
T C T T T T G T C A A T T A G G T C A C T - - - - -	269:23059:4074
- - - - - C A C T T C A A C C T C T T A T T G G A A G A T T A T C	269:23059:4074
- - - - - C A C T T C A A C C T C T T A T T G G A A G A T T A T C	97:24427:20792
- - - - - A C T T C A A C C T C T T A T T G G A A G A T T A T C	188:31952:1737
- - - - - C T T C A A C C T C T T A T T G G A A G A T T A T C	157:45607:9663
- - - - - C T T C A A C C T C T T A T T G G A A G A T T A T C	271:48926:6316
- - - - - C T T C A A C C T C T T A T T G G A A G A T T A T C	157:45607:9663
- - - - - C T T C A A C C T C T T A T T G G A A G A T T A T C	271:48926:6316
- - - - - T T C A A C C T C T T A T T G G A A G A T T A T C	25:19472:12305
- - - - - - - - - - - C	139:7453:11825

WT_3 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C	reference

(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 4 of 6

KDM6A_ko_1 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

[illegible]

KDM6A_ko_1 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T	C	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	reference									
NOT_COVERING (n=2)																																																					
T	C	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	83:12217:18859	
T	C	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	.	101:11853:2344

(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 5 of 6

KDM6A_ko_2 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

[illegible]

KDM6A_ko_2 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C reference

(B) KDM6A locus, chrX:44,961,342-44,961,386 — sheet 6 of 6

KDM6A_ko_3 RNA -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A_ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	A	A	C	C	T	C	T	T	A	T	T	G	G	A	A	G	A	T	T	A	T	C	reference
NOT_COVERING (n=8)																																													
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	T	A	-	A	G	A	G	G	A	T	C	C	G	A	A	G	A	T	T	A	T	C	48:41216:26813
T	C	T	T	T	T	G	T	C	A	A	T	T	A	G	G	T	C	A	C	T	T	C	T	A	-	A	G	A	G	G	A	T	C	C	G	A	A	G	A	T	T	A	T	C	298:10186:4571
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	G	A	A	G	A	T	T	A	T	C	128:8794:13762
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	G	A	A	G	A	T	T	A	T	C	87:22763:24411
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	G	A	A	G	A	T	T	A	T	C	48:41216:26813
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	G	A	A	G	A	T	T	A	T	C	128:8794:13762
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	G	A	A	G	A	T	T	A	T	C	87:22763:24411
-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	-	A	G	A	T	T	A	T	C	298:10186:4571	

KDM6A_ko_3 ATAC -- chrX:44,961,342-44,961,386 -- anchor chrX:44,961,364 (edit highlighted)
Sane WT vs Sane KDM6A ko (DESeq2 Wald test, size-factor normalized): log2FC=-5.00, padj=0.019 (*)

T C T T T T G T C A A T T A G G T C A C T T C A A C C T C T T A T T G G A A G A T T A T C reference