

1 SETDB1

This gene encodes a histone methyltransferase which regulates histone methylation, gene silencing, and transcriptional repression. This gene has been identified as a target for treatment in Huntington Disease, given that gene silencing and transcription dysfunction likely play a role in the disease pathogenesis. Alternatively spliced transcript variants of this gene have been described. Histone methyltransferase that specifically trimethylates 'Lys-9' of histone H3. H3 'Lys-9' trimethylation represents a specific tag for epigenetic transcriptional repression by recruiting HP1 (CBX1, CBX3 and/or CBX5) proteins to methylated histones. Mainly functions in euchromatin regions, thereby playing a central role in the silencing of euchromatic genes. H3 'Lys-9' trimethylation is coordinated with DNA methylation. Probably forms a complex with MBD1 and ATF7IP that represses transcription and couples DNA methylation and histone 'Lys-9' trimethylation. Its activity is dependent on MBD1 and is heritably maintained through DNA replication by being recruited by CAF-1. SETDB1 is targeted to histone H3 by TRIM28/TIF1B, a factor recruited by KRAB zinc-finger proteins. Gene expression is slightly upregulated in Hu7 23 hours after Ebola infection, but not in bat. The transcript may in some cases be fused with the transcript of the downstream gene coding for CERS2.

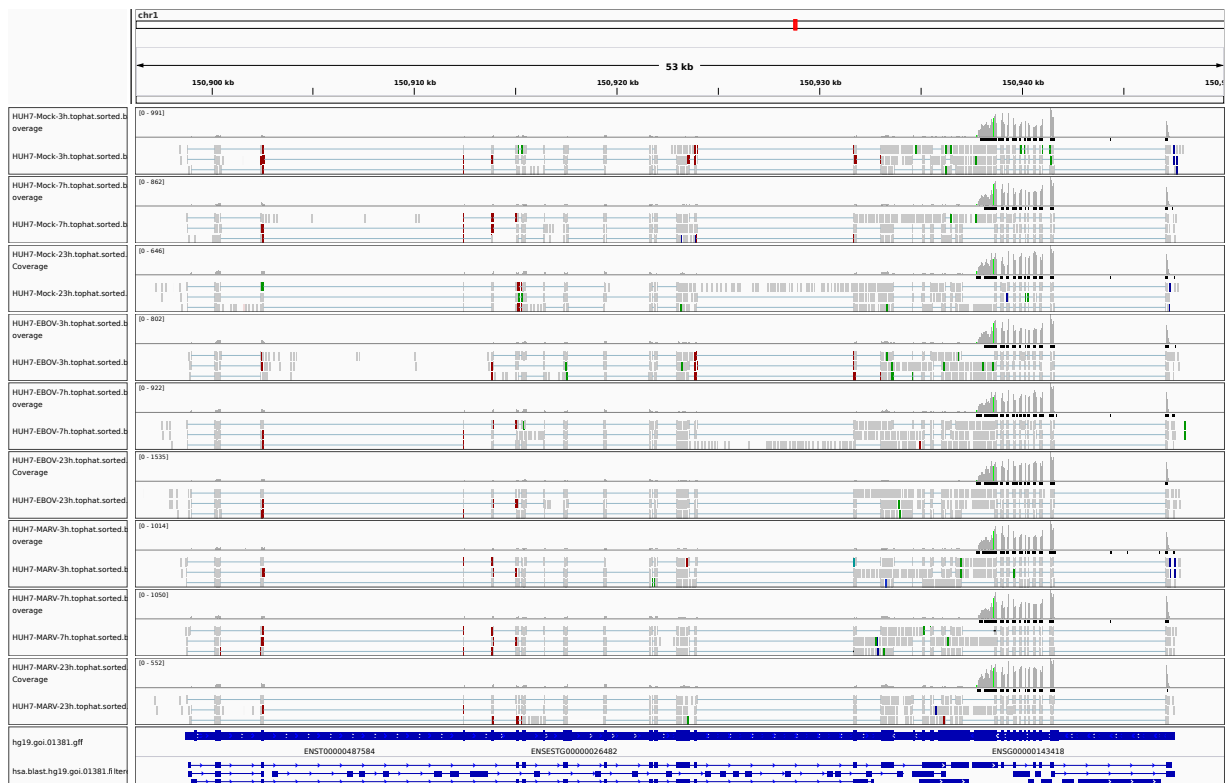


Figure 1: IGV Genome Browser screenshot of gene SETDB1.

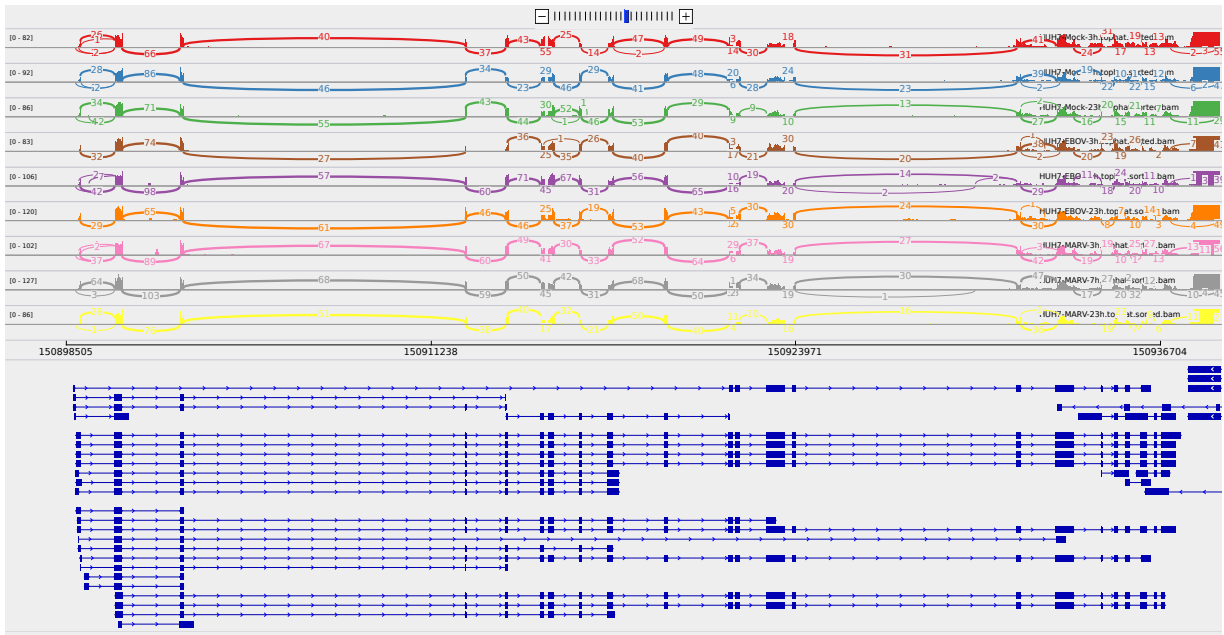


Figure 2: Sashimi plot of gene SETDB1.

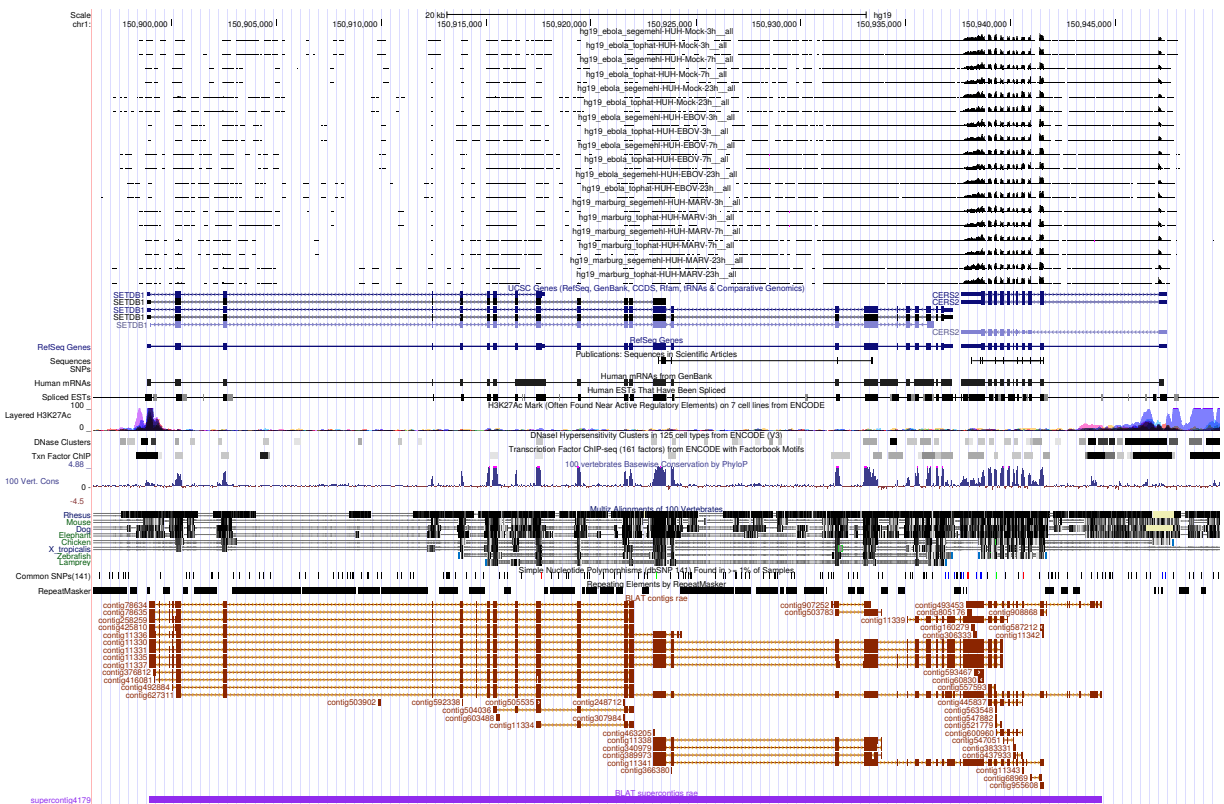


Figure 3: UCSC Genome Browser screenshot of gene SETDB1.