

1 ADAMTS2

This gene encodes a member of the ADAMTS (a disintegrin and metalloproteinase with thrombospondin motifs) protein family. Members of the family share several distinct protein modules, including a propeptide region, a metalloproteinase domain, a disintegrin-like domain, and a thrombospondin type 1 (TS) motif. Individual members of this family differ in the number of C-terminal TS motifs, and some have unique C-terminal domains. The enzyme encoded by this gene excises the N-propeptide of type I, type II and type V procollagens. Mutations in this gene cause Ehlers-Danlos syndrome type VIIC, a recessively inherited connective-tissue disorder. Alternative splicing results in multiple transcript variants.

No expression in human and in the bat homolog. Something is expressed slightly directly 5' to the human gene (not annotated as a gene). No gene-expression plot was done for the gene, because there is nothing to see and it covers about 240kb.



Figure 1: UCSC Genome Browser screenshot of gene ADAMTS2.

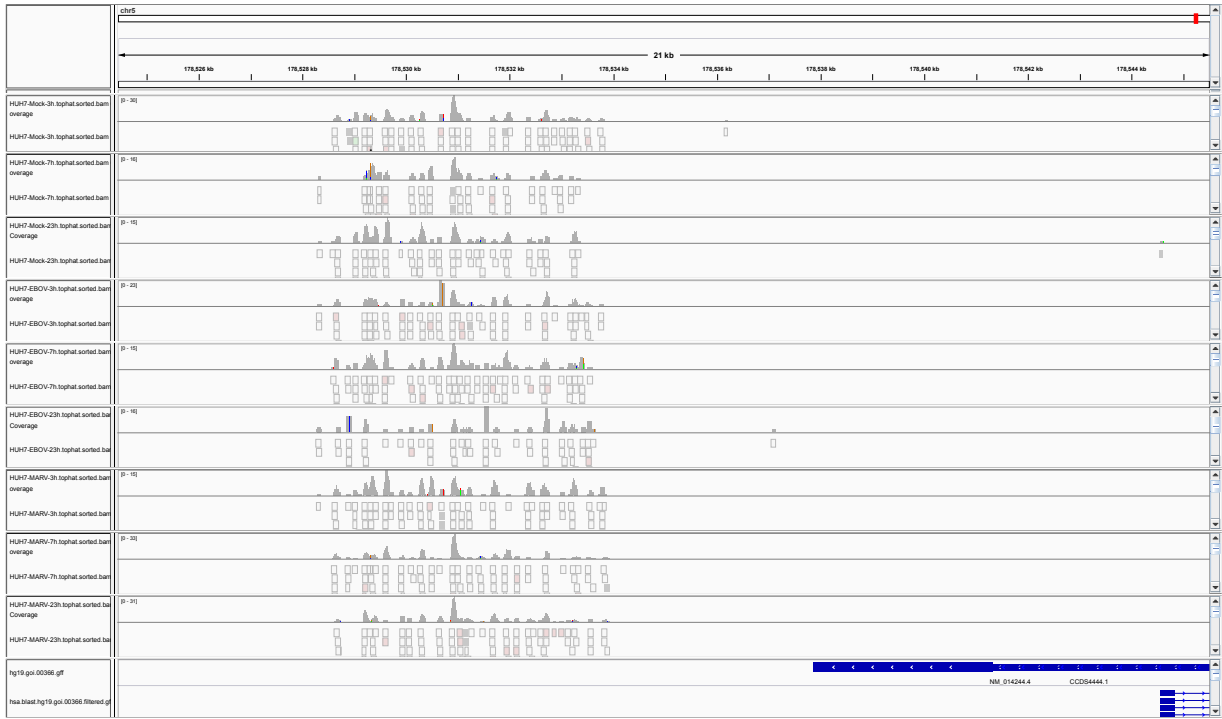


Figure 2: IGV Genome Browser screenshot of region directly 5' of gene ADAMTS2 with some expression visible.