1 TGFBR1

The protein encoded by this gene forms a heteromeric complex with type II TGF-beta receptors when bound to TGF-beta, transducing the TGF-beta signal from the cell surface to the cytoplasm. The encoded protein is a serine/threonine protein kinase. Mutations in this gene have been associated with Loeys-Dietz aortic aneurysm syndrome (LDAS). Multiple transcript variants encoding different isoforms have been found for this gene.

This gene is somewhat differentially expressed, although not clearly so. It is highly expressed in bat in the single exon which matched the human genome well. There is also some expression in human, in particular in the 3'UTR.

Figure 1: IGV Genome Browser screenshot of gene TGFBR1.

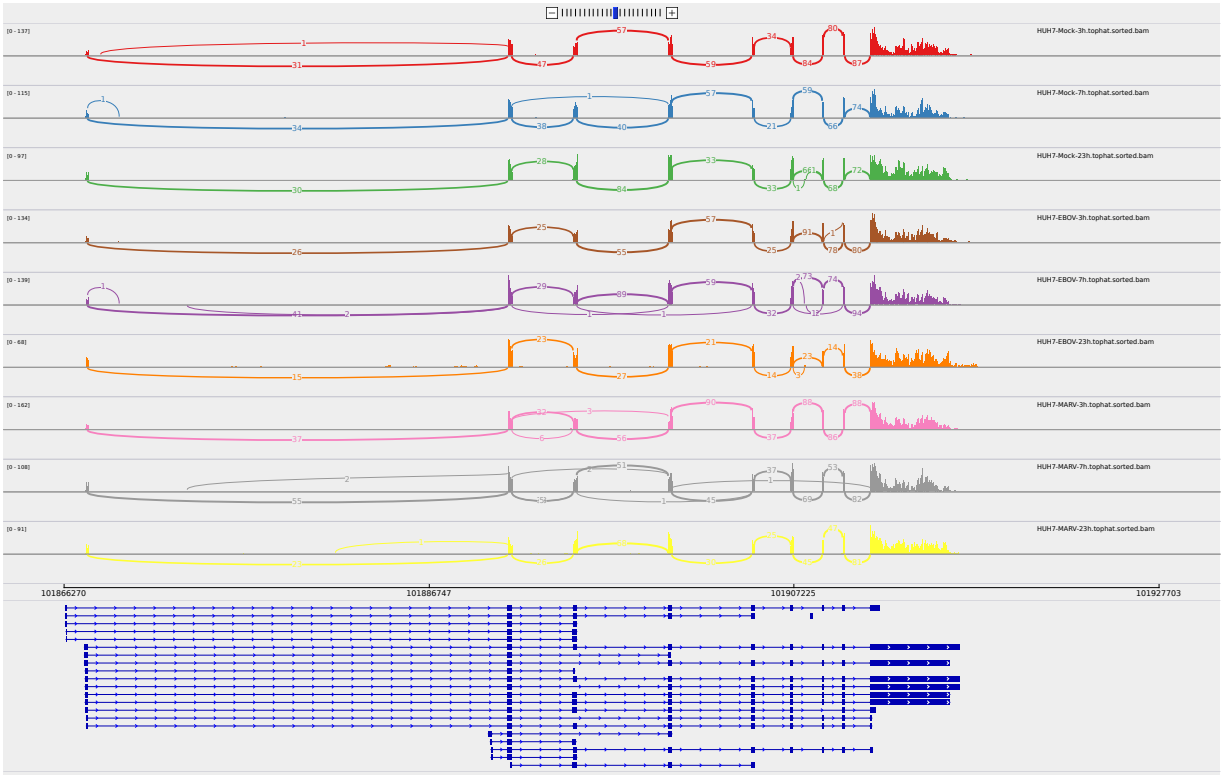

Figure 2: Sashimi plot of gene TGFBR1.

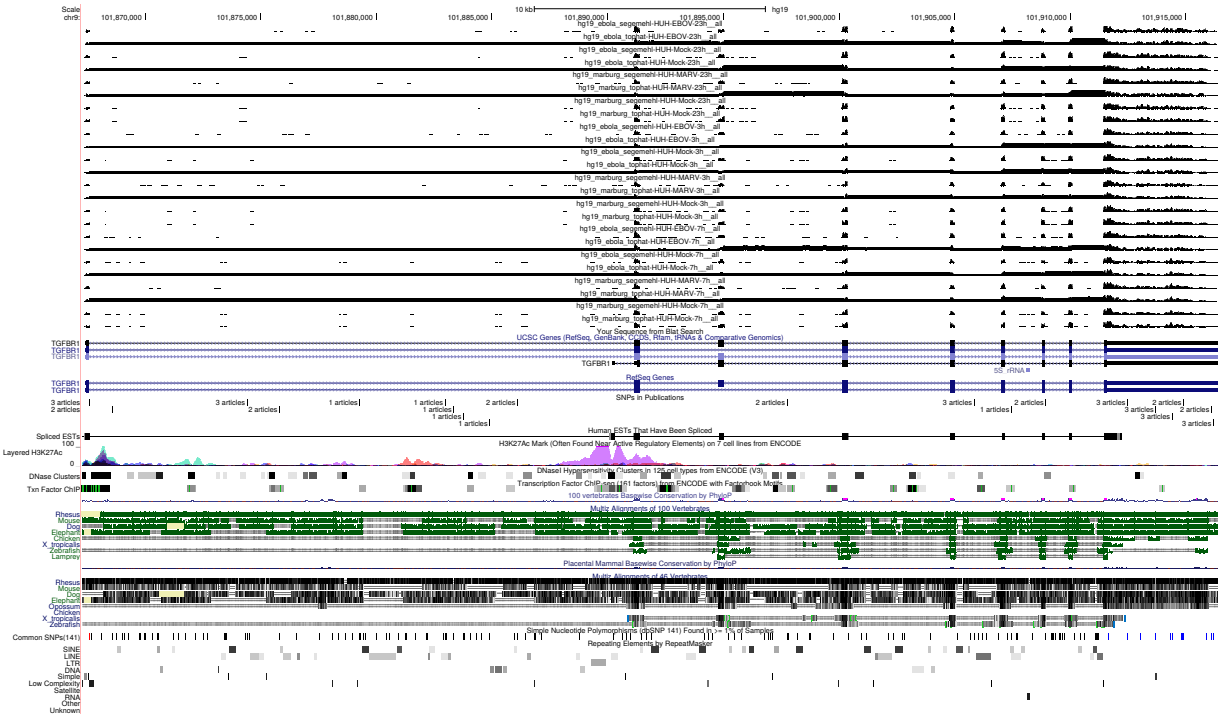

Figure 3: UCSC Genome Browser screenshot of gene TGFBR1.