

1 CAV3

This gene encodes a caveolin family member, which functions as a component of the caveolae plasma membranes found in most cell types. Caveolin proteins are proposed to be scaffolding proteins for organizing and concentrating certain caveolin-interacting molecules. Mutations identified in this gene lead to interference with protein oligomerization or intra-cellular routing, disrupting caveolae formation and resulting in Limb-Girdle muscular dystrophy type-1C (LGMD-1C), hyperCKemia or rippling muscle disease (RMD). Alternative splicing has been identified for this locus, with inclusion or exclusion of a differentially spliced intron.

The gene is not expressed.

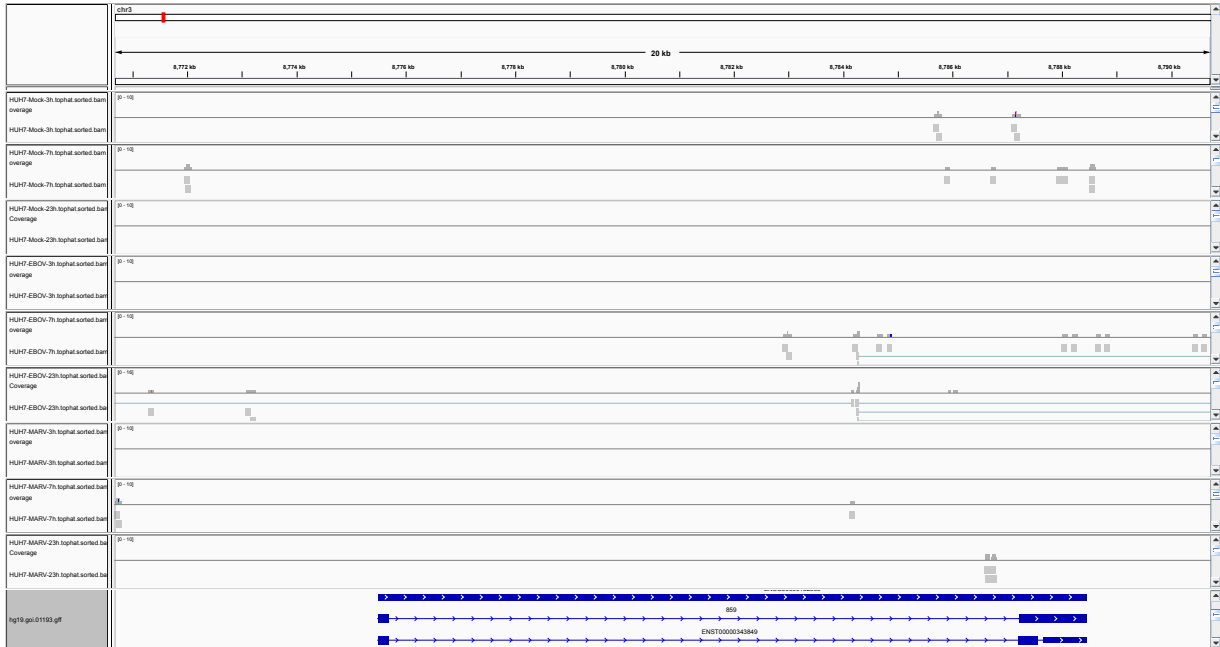


Figure 1: IGV Genome Browser screenshot of gene CAV3.

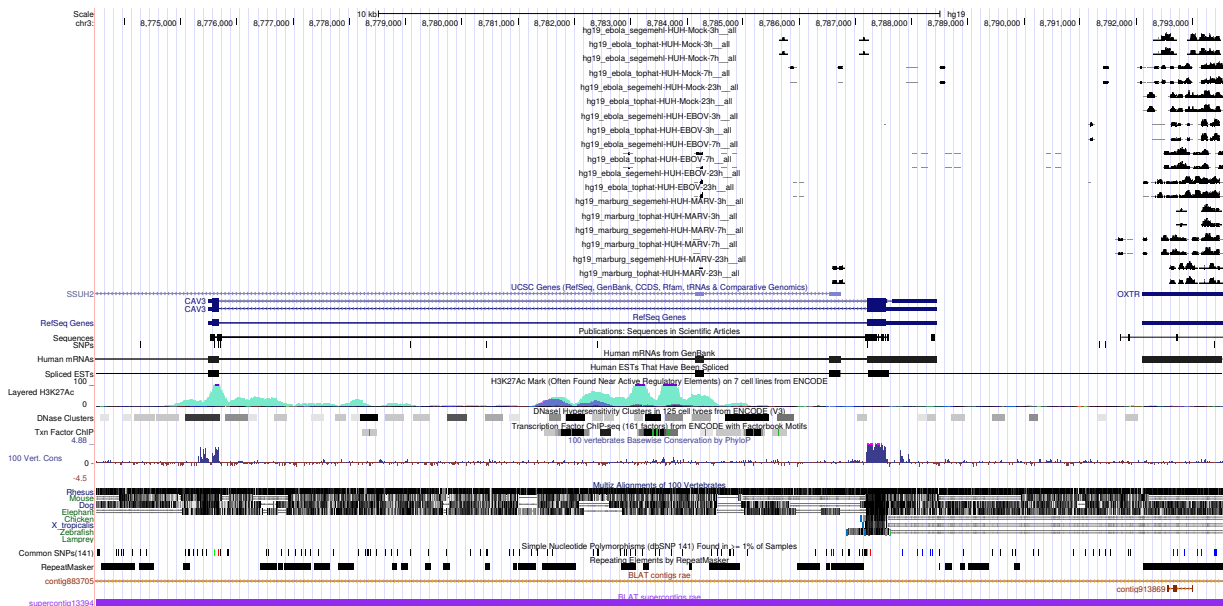


Figure 2: UCSC Genome Browser screenshot of gene CAV3.