

1 SHH

This gene encodes a protein that is instrumental in patterning the early embryo. It has been implicated as the key inductive signal in patterning of the ventral neural tube, the anterior-posterior limb axis, and the ventral somites. Of three human proteins showing sequence and functional similarity to the sonic hedgehog protein of *Drosophila*, this protein is the most similar. The protein is made as a precursor that is autocatalytically cleaved; the N-terminal portion is soluble and contains the signalling activity while the C-terminal portion is involved in precursor processing. More importantly, the C-terminal product covalently attaches a cholesterol moiety to the N-terminal product, restricting the N-terminal product to the cell surface and preventing it from freely diffusing throughout the developing embryo. Defects in this protein or in its signalling pathway are a cause of holoprosencephaly (HPE), a disorder in which the developing forebrain fails to correctly separate into right and left hemispheres. HPE is manifested by facial deformities. It is also thought that mutations in this gene or in its signalling pathway may be responsible for VACTERL syndrome, which is characterized by vertebral defects, anal atresia, tracheoesophageal fistula with esophageal atresia, radial and renal dysplasia, cardiac anomalies, and limb abnormalities. Additionally, mutations in a long range enhancer located approximately 1 megabase upstream of this gene disrupt limb patterning and can result in preaxial polydactyly. The gene is upregulated in human EBOV. The first 3'UTR exon is extended to the second one (which are annotated as being alternative). No homologs in bat were found.

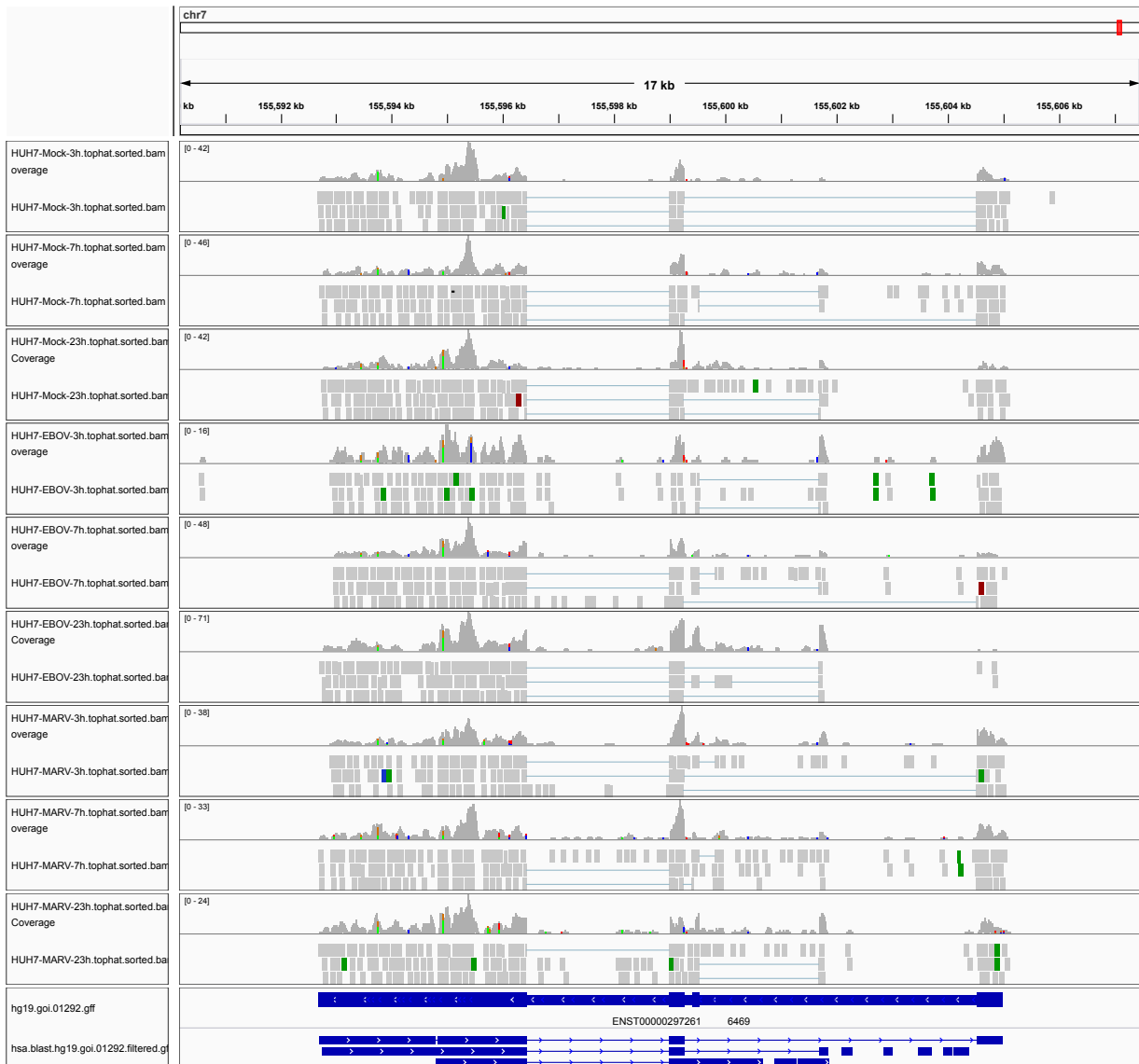


Figure 1: IGV Genome Browser screenshot of gene SHH.

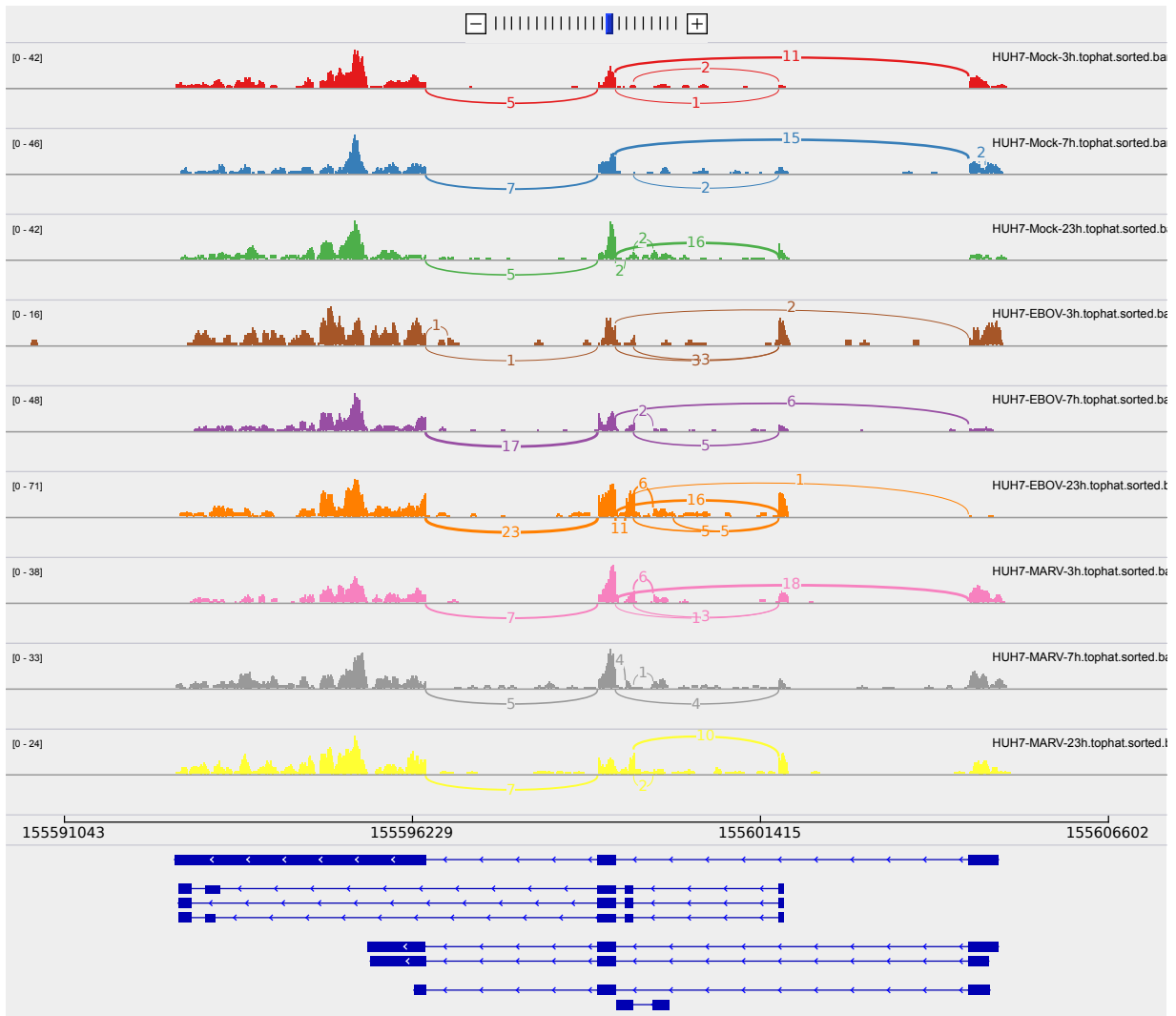


Figure 2: Sashimi plot of gene SHH.

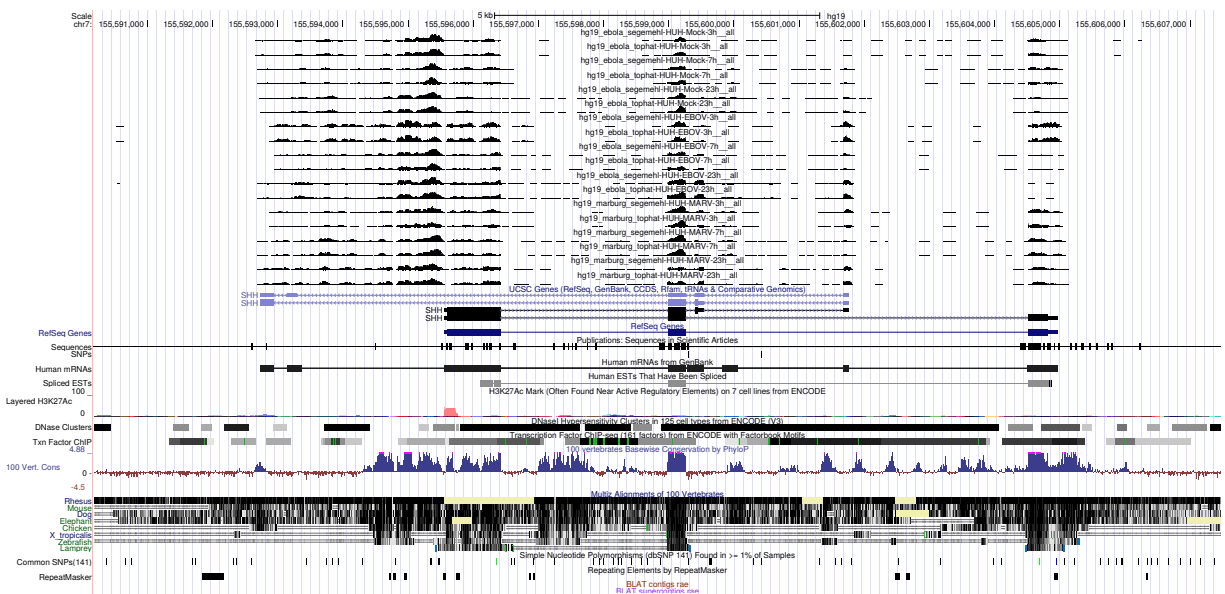


Figure 3: UCSC Genome Browser screenshot of gene SHH.