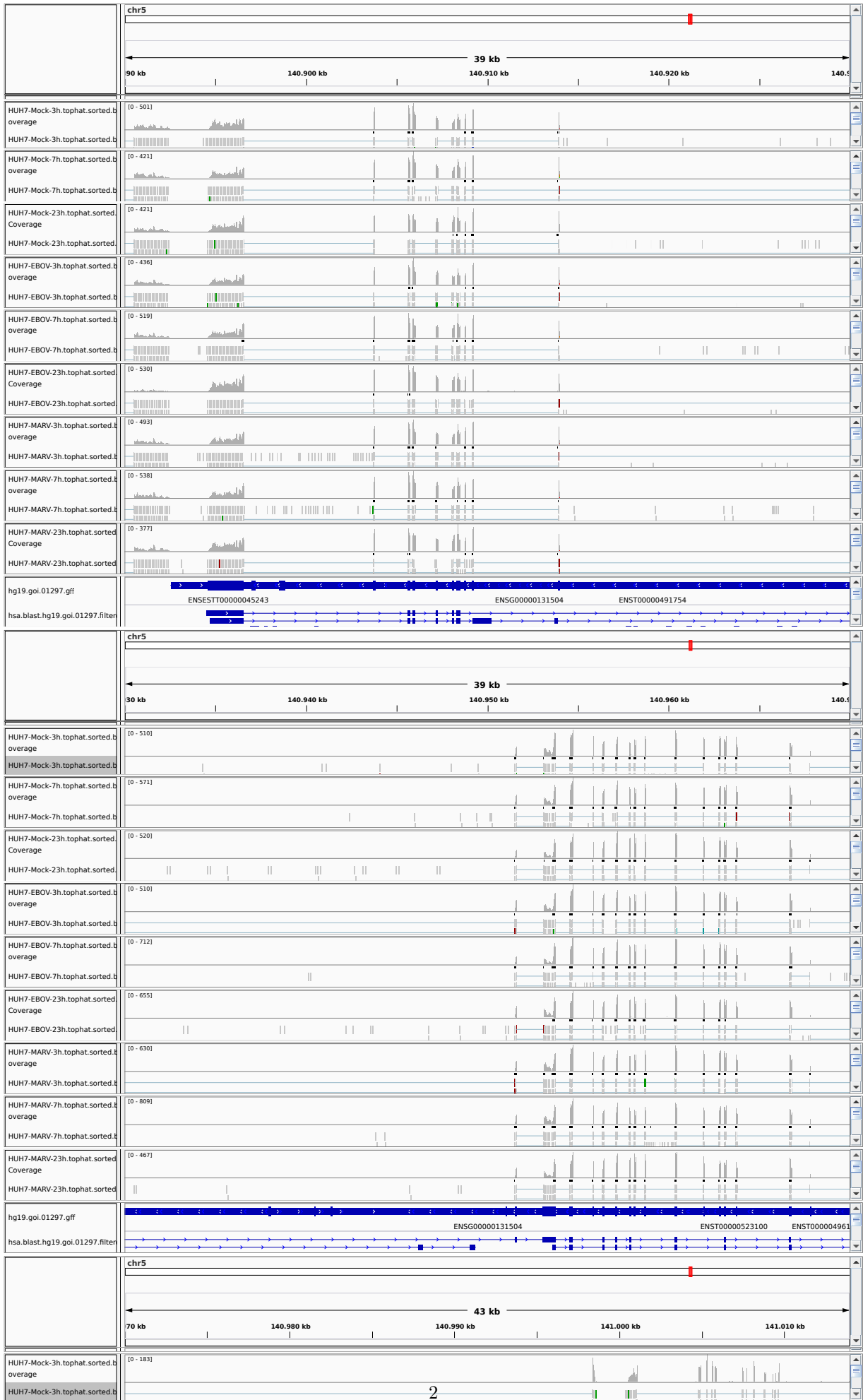


# 1 DIAPH1

This gene is a homolog of the *Drosophila* diaphanous gene, and has been linked to autosomal dominant, fully penetrant, nonsyndromic sensorineural progressive low-frequency hearing loss. Actin polymerization involves proteins known to interact with diaphanous protein in *Drosophila* and mouse. It has therefore been speculated that this gene may have a role in the regulation of actin polymerization in hair cells of the inner ear. Alternatively spliced transcript variants encoding distinct isoforms have been found for this gene. The DIAPH1 is known to be associated in the CDC42 pathway and is up-regulated in human cells infected with ebola and Marburg-virus. Also the same expression pattern is observed in the surrounding genes HDAC3 (upstream) and PCDHGA1 (downstream).



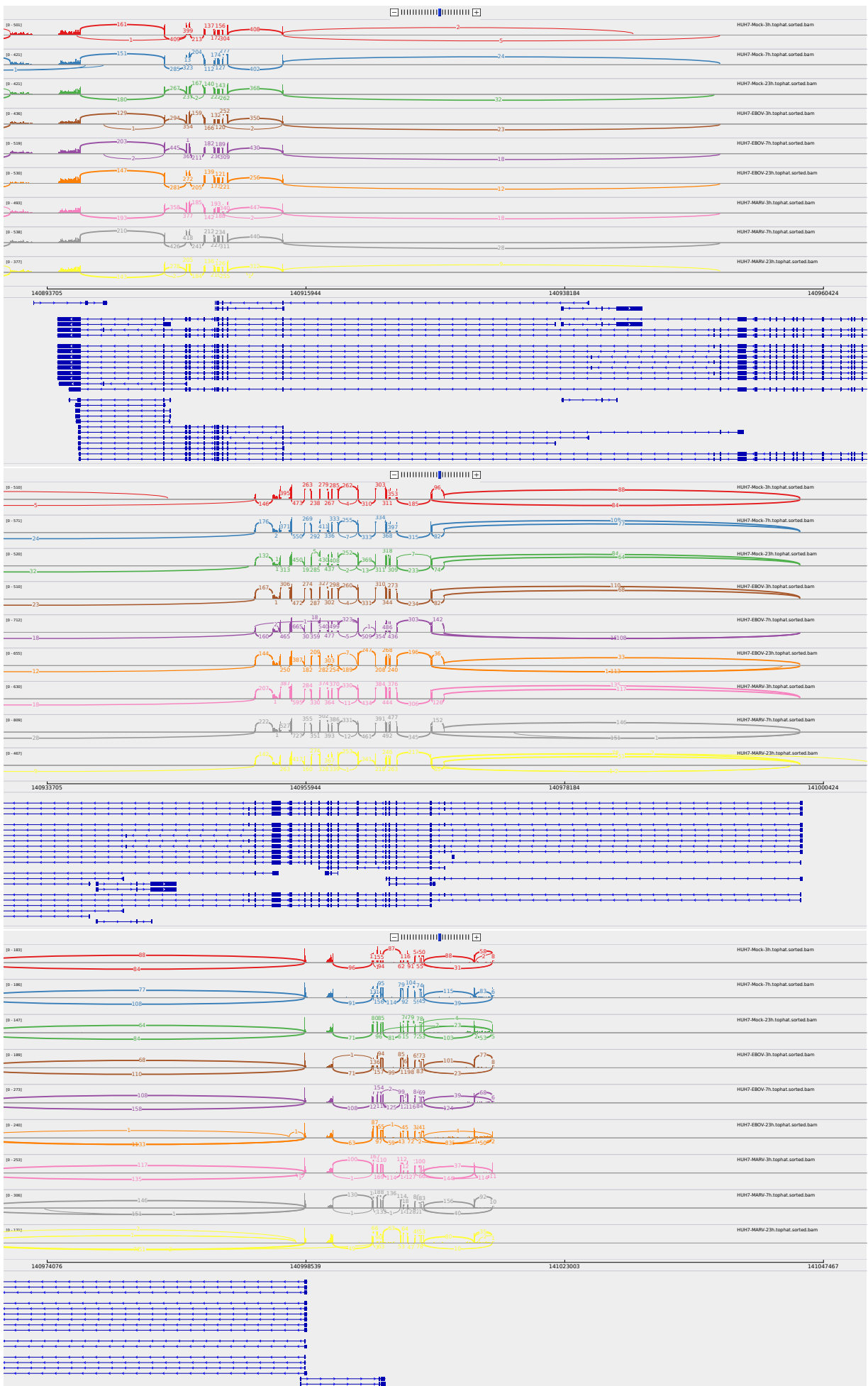


Figure 2: Sashimi plot of gene DIAPH1.

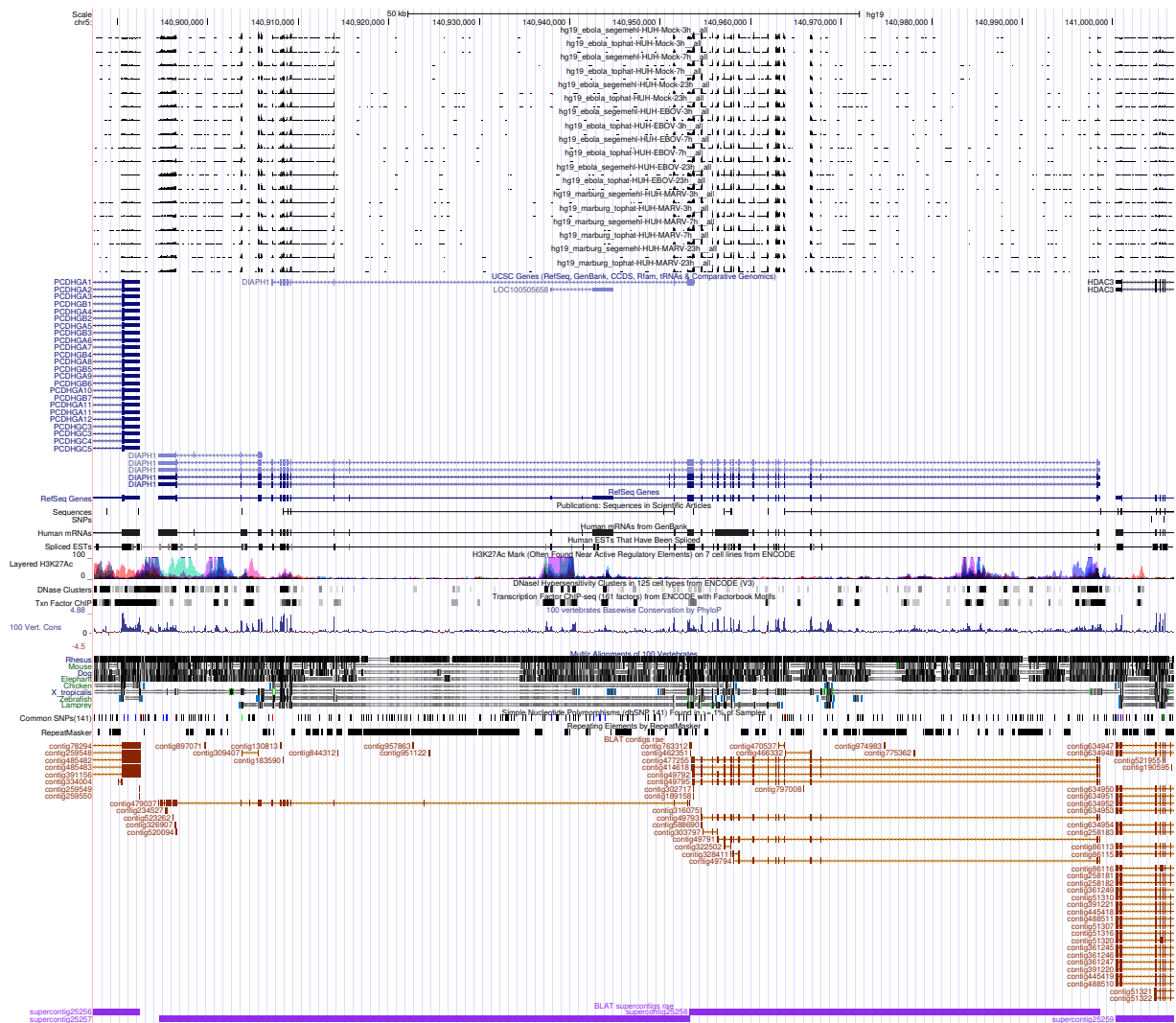


Figure 3: UCSC Genome Browser screenshot of gene *DIAPH1*.