

# 1 INPP5E

The protein encoded by this gene is an inositol 1,4,5-trisphosphate (InsP3) 5-phosphatase. InsP3 5-phosphatases hydrolyze Ins(1,4,5)P3, which mobilizes intracellular calcium and acts as a second messenger mediating cell responses to various stimulation. Studies of the mouse counterpart suggest that this protein may hydrolyze phosphatidylinositol 3,4,5-trisphosphate and phosphatidylinositol 3,5-bisphosphate on the cytoplasmic Golgi membrane and thereby regulate Golgi-vesicular trafficking. Mutations in this gene cause Joubert syndrome; a clinically and genetically heterogenous group of disorders characterized by midbrain-hindbrain malformation and various associated ciliopathies that include retinal dystrophy, nephronophthisis, liver fibrosis and polydactyly.

This gene is not differentially expressed in all probes.

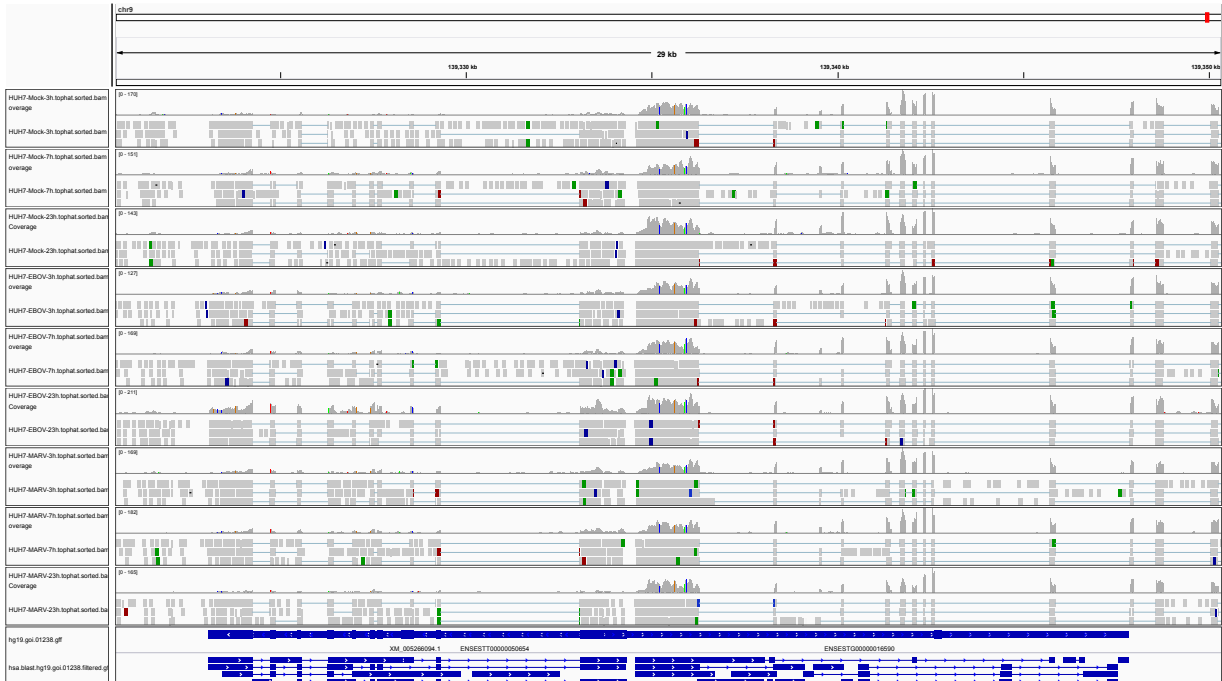


Figure 1: IGV Genome Browser screenshot of gene INPP5E.

Figure 2: Sashimi plot of gene INPP5E.

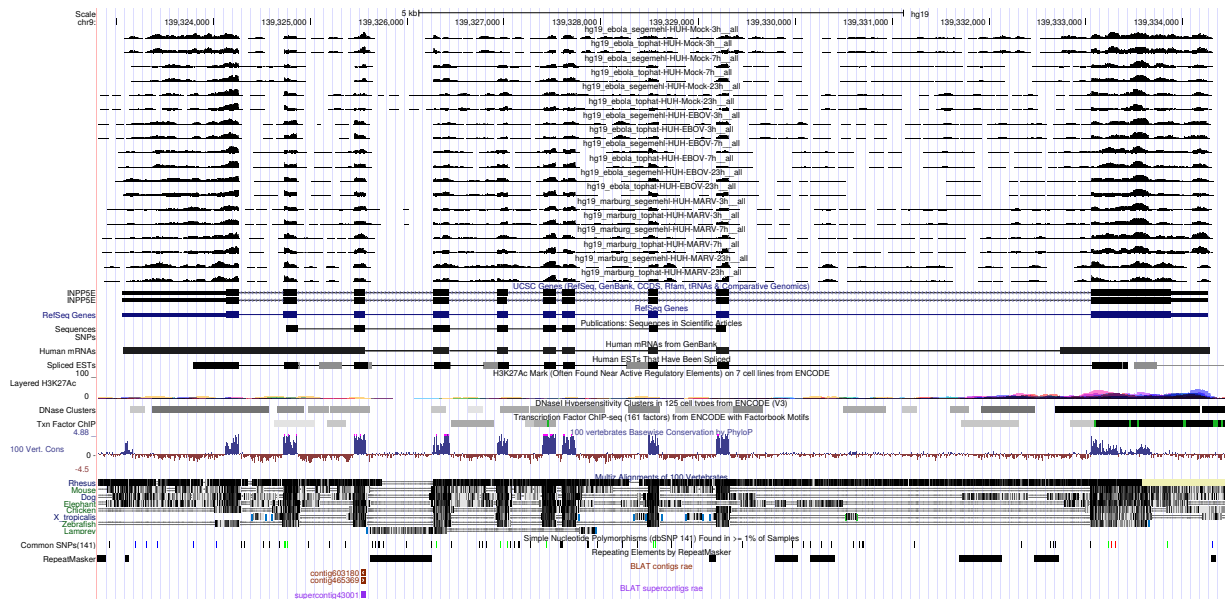


Figure 3: UCSC Genome Browser screenshot of gene INPP5E.