# 1 NPC1

This gene encodes a large protein that resides in the limiting membrane of endosomes and lysosomes and mediates intracellular cholesterol trafficking via binding of cholesterol to its N-terminal domain. It is predicted to have a cytoplasmic C-terminus, 13 transmembrane domains, and 3 large loops in the lumen of the endosome - the last loop being at the N-terminus. This protein transports low-density lipoproteins to late endosomal/lysosomal compartments where they are hydrolized and released as free cholesterol. Defects in this gene cause Niemann-Pick type C disease, a rare autosomal recessive neurodegenerative disorder characterized by over accumulation of cholesterol and glycosphingolipids in late endosomal/lysosomal compartments.

The NPC1 appears to be essential for Ebola and Marburg virus infection **?**. The NPC1 is moderately expressed in human samples and not detectable in bat samples. In human samples the expression is upregulated over time. Interestingly, on the opposite strand the open reading frame 8 on chromosome 18 (C18orf8) seems to be co-regulated. Additionally the C18orf8 shows unique alternative splicing events during Ebola and Marburg virus infection.

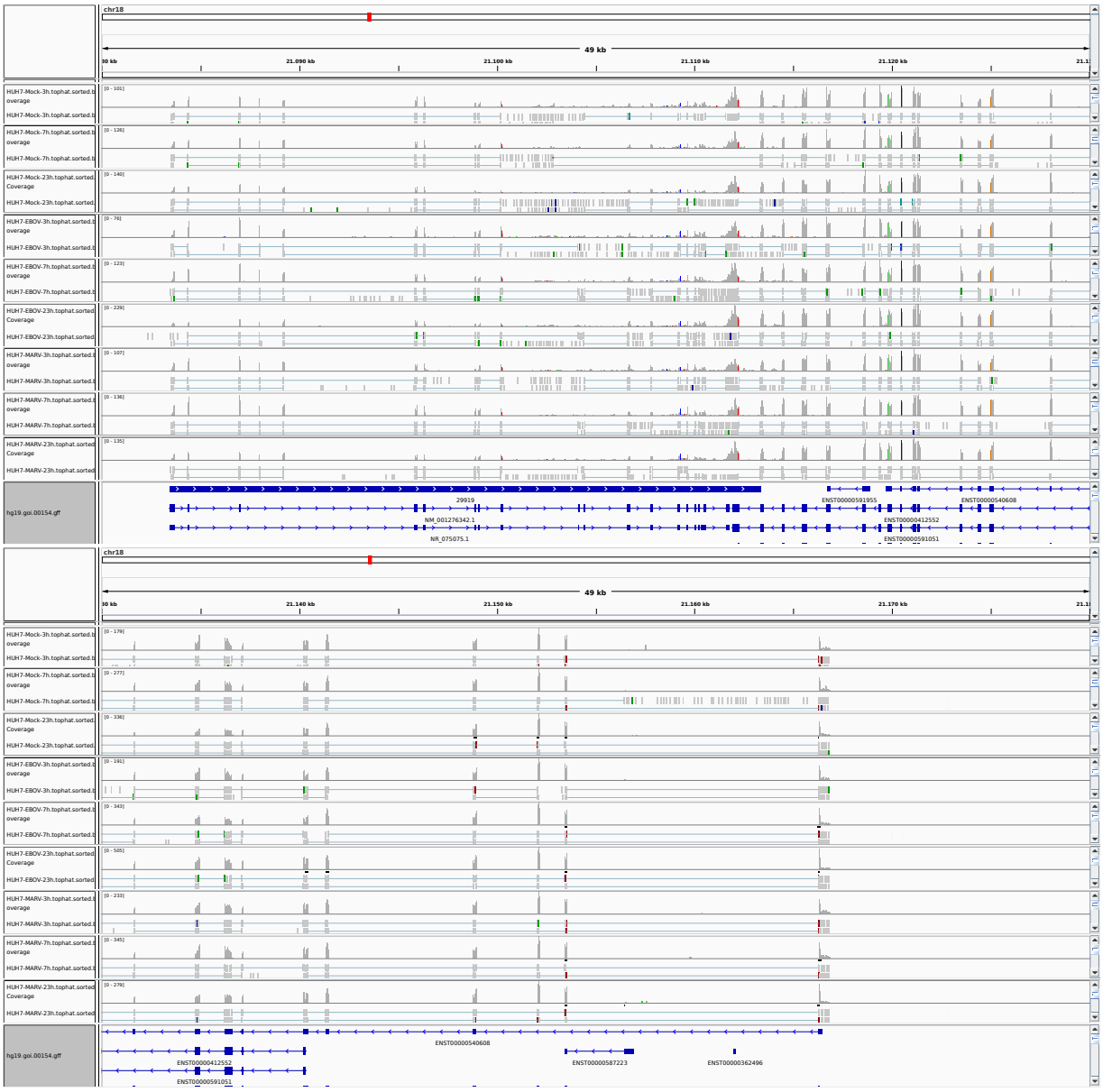


Figure 1: IGV Genome Browser screenshot of gene NPC1.

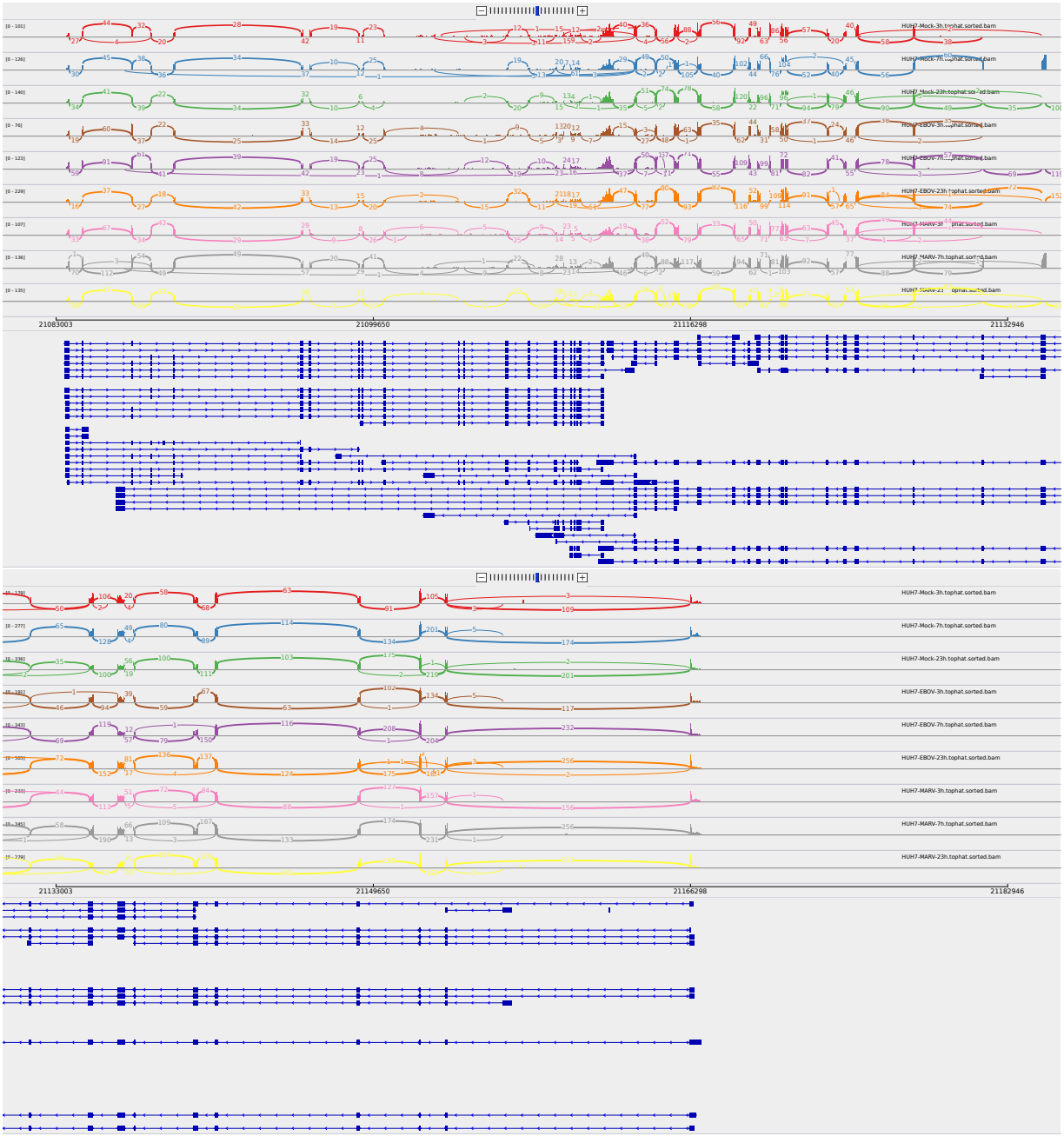


Figure 2: Sashimi plot of gene NPC1.

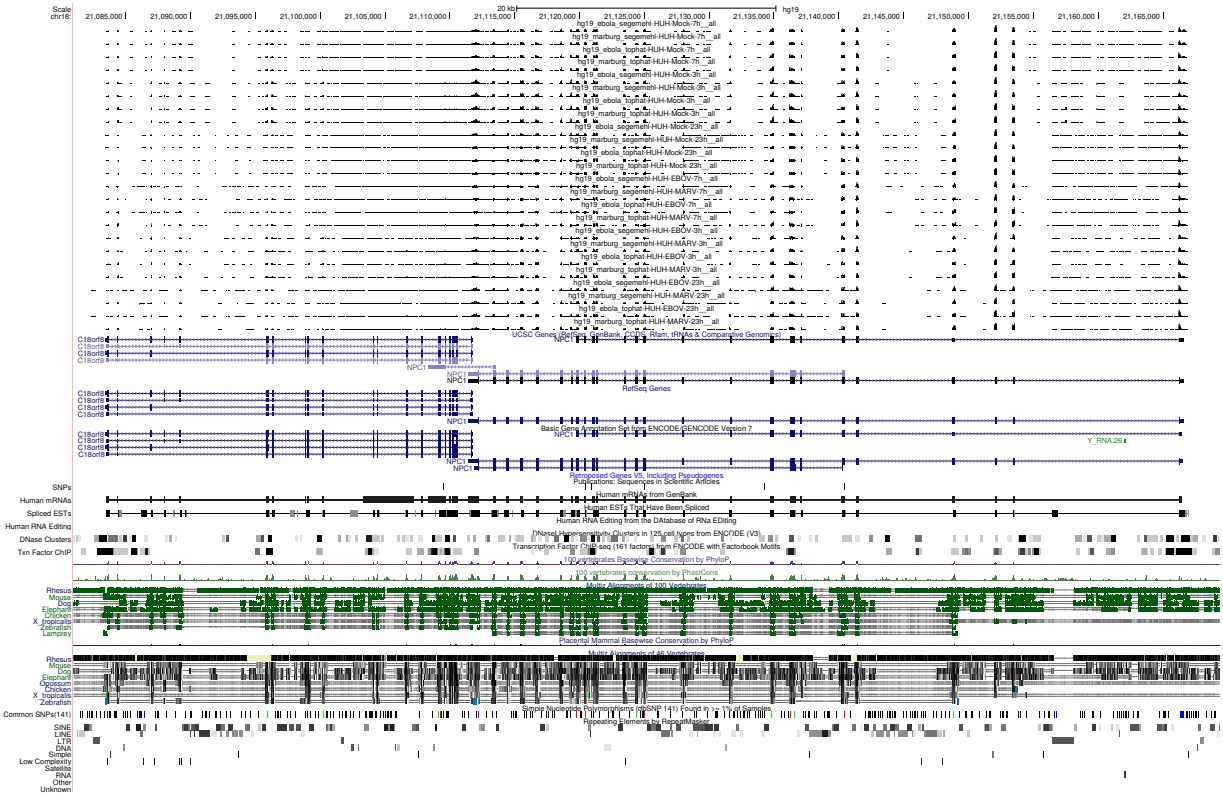

Figure 3: UCSC Genome Browser screenshot of gene NPC1.

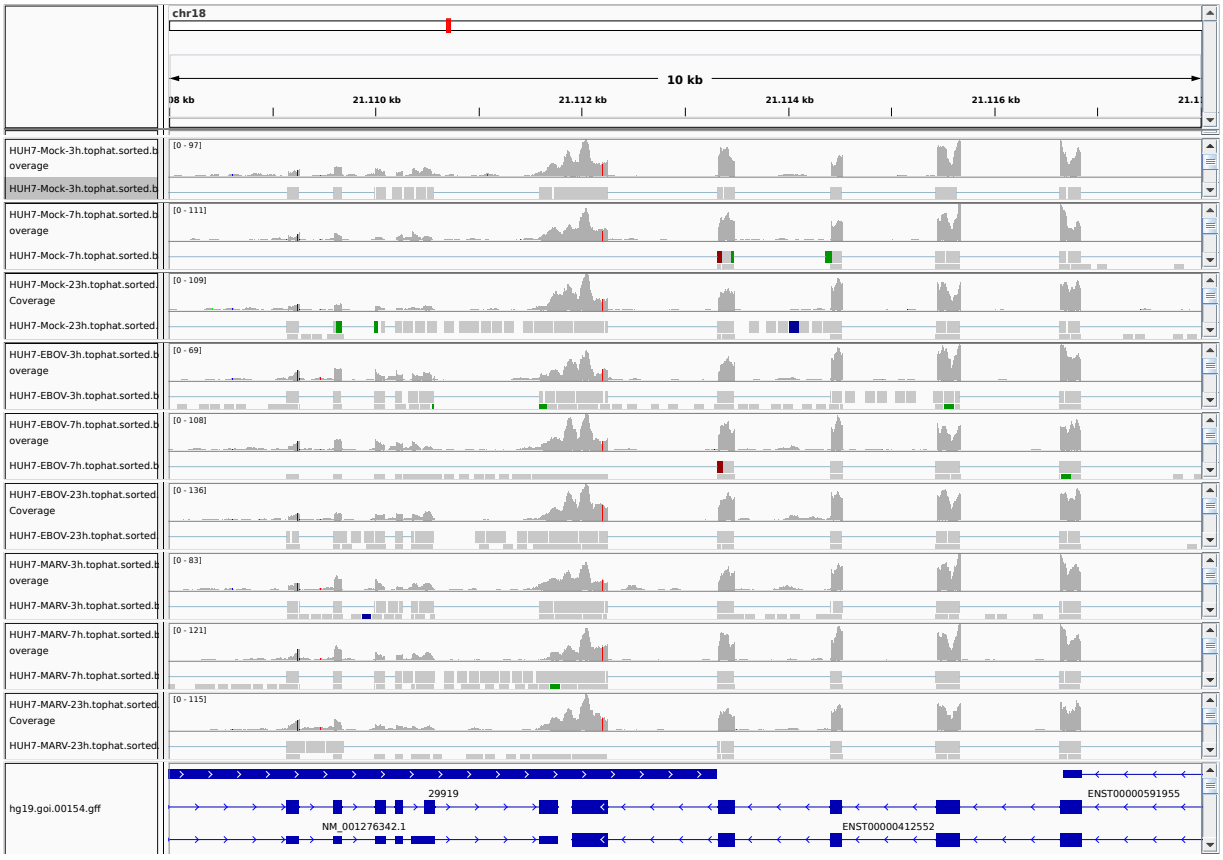


Figure 4: The ends of NPC1 (right) and the open reading frame C18orf8 (left) on the opposite strand laying next to each other. Note the same scale of read counts suggesting coregulation of both.