1 ATXN2

The autosomal dominant cerebellar ataxias (ADCA) are a heterogeneous group of neurodegenerative disorders characterized by progressive degeneration of the cerebellum, brain stem and spinal cord. Clinically, ADCA has been divided into three groups: ADCA types I-III. Defects in this gene are the cause of spinocerebellar ataxia type 2 (SCA2). SCA2 belongs to the autosomal dominant cerebellar ataxias type I (ADCA I) which are characterized by cerebellar ataxia in combination with additional clinical features like optic atrophy, ophthalmoplegia, bulbar and extrapyramidal signs, peripheral neuropathy and dementia. SCA2 is caused by expansion of a CAG repeat in the coding region of this gene. This locus has been mapped to chromosome 12, and it has been determined that the diseased allele contains 37-50 CAG repeats, compared to 17-29 in the normal allele. Longer expansions result in earlier onset of the disease.

Gene is upregulated upon Ebola but not Marburg virus infection in human, and in both, Marburg and Ebola virus infection in bat.

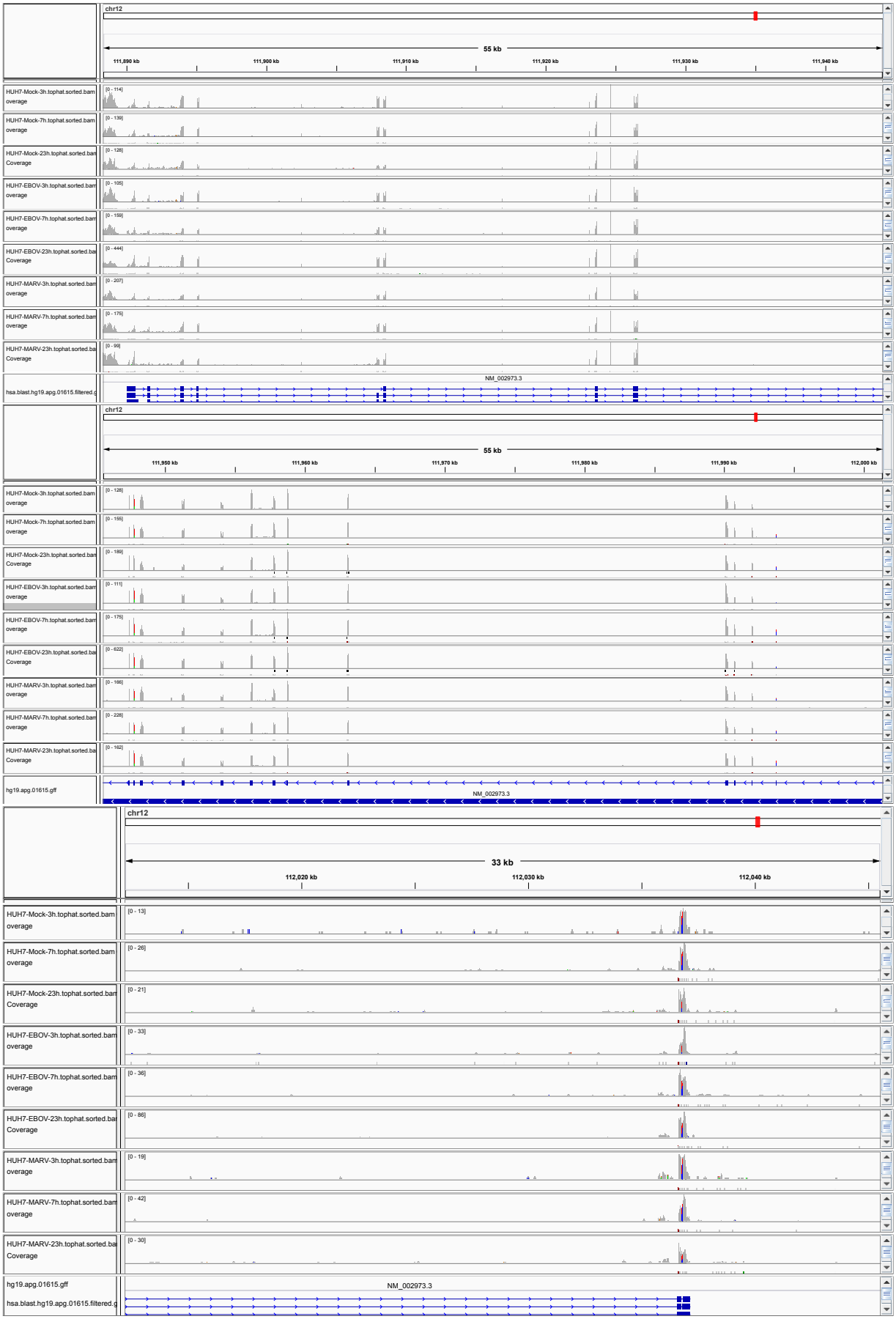

Figure 1: IGV Genome Browser screenshot of gene ATXN2.

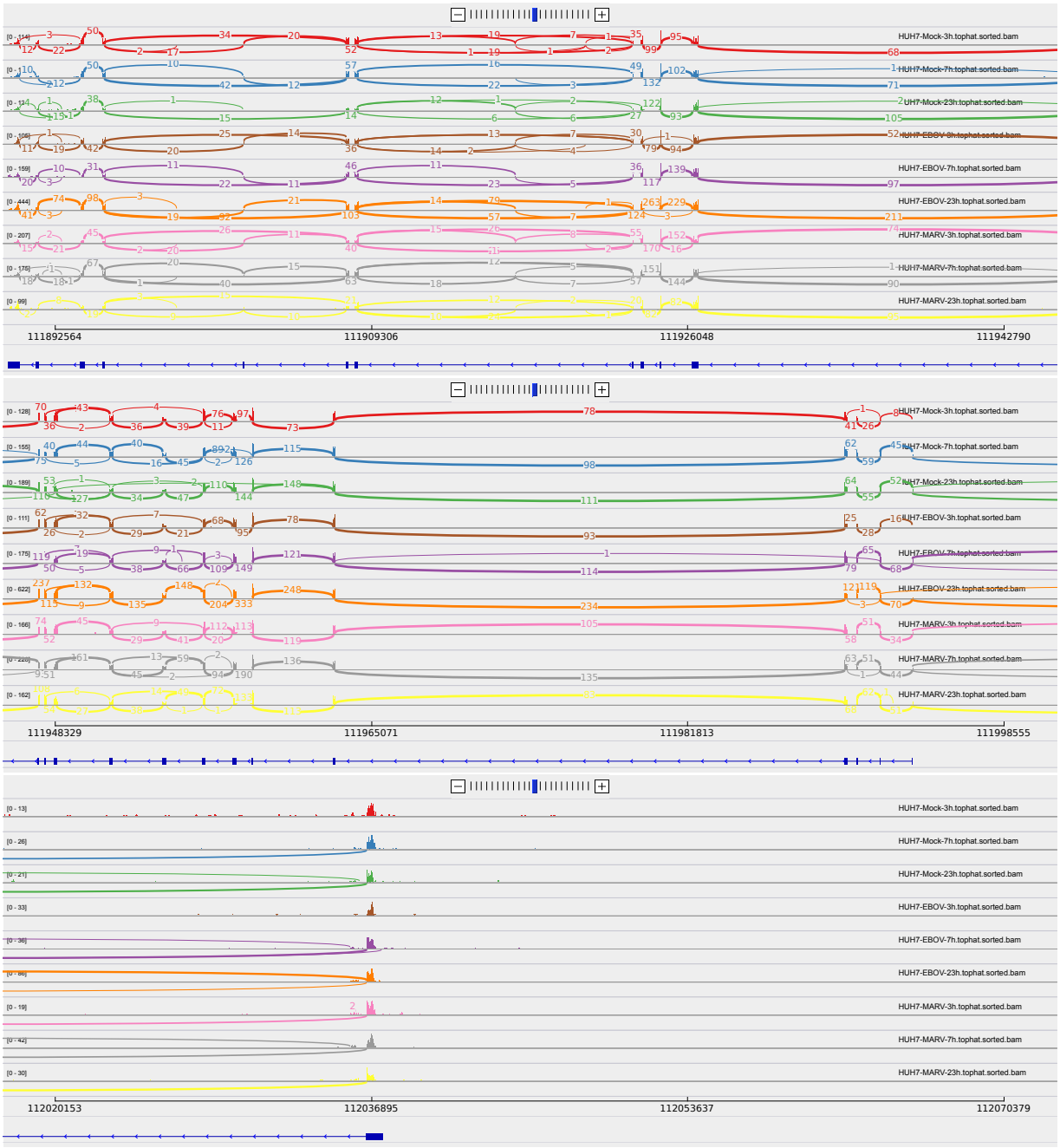

Figure 2: Sashimi plot of gene ATXN2.

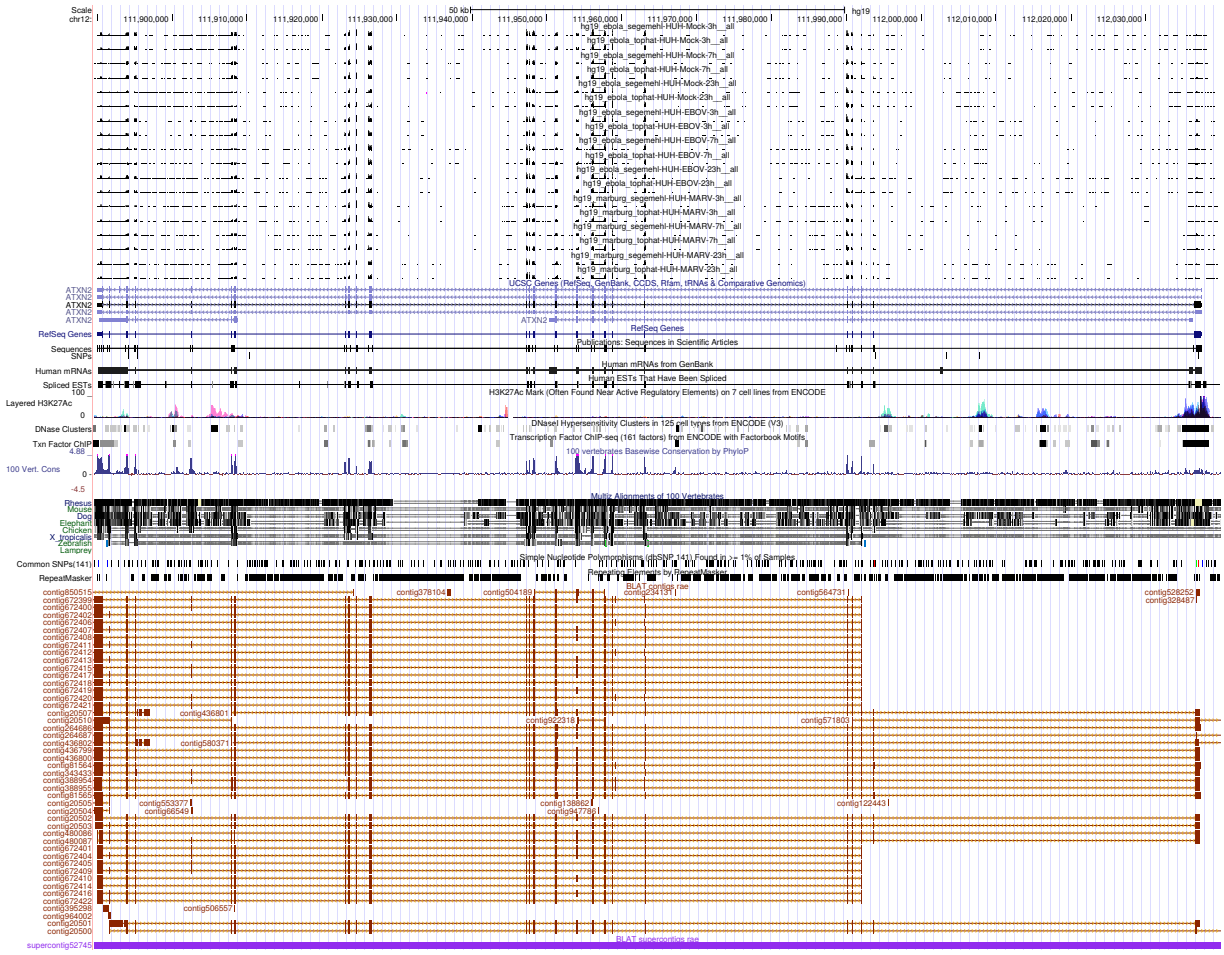

Figure 3: UCSC Genome Browser screenshot of gene ATXN2.