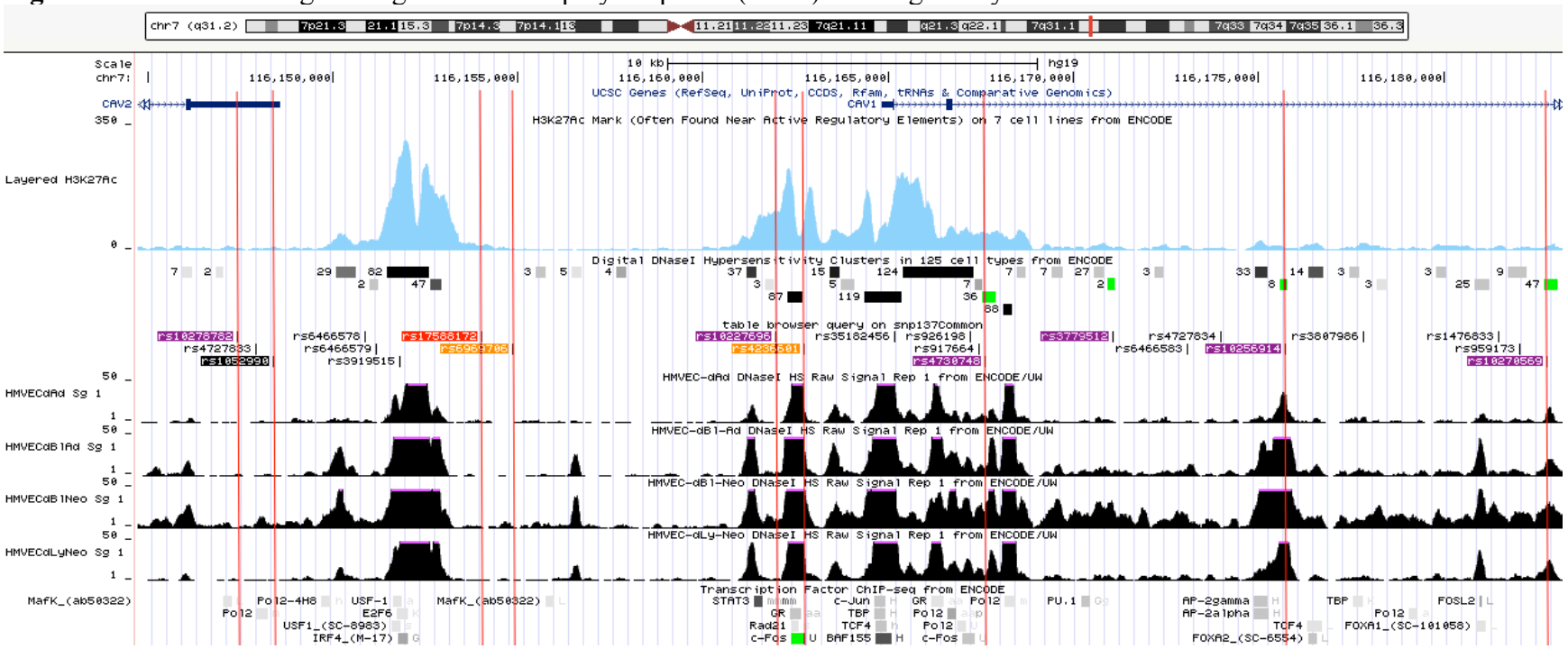
Figure 4: *CAV1/CAV2* region single nucleotide polymorphism (SNPs) with regulatory information.



The *CAV1/CAV2* intragenic region including the ten significantly associated SNPs and regulatory elements are shown using the UCSC genome browser *CAV1/CAV2* region *(*http://genome.ucsc.edu, Accessed April 2, 2013*)*. The SNPs that are significant overall, in women and in cases with paracentral visual field loss are highlighted in orange. SNPs that are significant only overall and in women are highlighted in purple. SNPs that are significant only overall and in cases with paracentral visual field (VF) loss are highlighted in red, and the SNP that is significant only overall is highlighted in black. Two of the SNPs lie in the 3' untranslated region (UTR) of *CAV2* (rs1052990 and rs10278782), four SNPs are in the intergenic region between *CAV1* and *CAV2*, and three SNPs are in intron 2 of *CAV1*. The red vertical lines orient the SNPs with their position relative to the regulatory elements present in this region. H3K27Ac histone marks in human umbilical vascular endothelial cells (HUVEC) (typically found in genomic regions with regulatory activity) are indicated by the blue peaks. DNaseI hypersensitivity sites (also indicating active regulatory sites) are identified as rectangles directly below the H3K27Ac blue peaks. The numbers next to each rectangle indicate the number of cells (from a total of 125) where the DNaseI site is active. Three of the associated SNPs (rs10256914, rs10270569 and rs4730748) are located in a DNaseI sites and each of these are highlighted in green. There were four cell types in common among the DNaseI sites that contain these 3 SNPs all of which are vascular cell lines: HMVEC-dAd (adult dermal microvascular endothelial cells), HMVEC-dBl-Ad (adult blood microvascular endothelial cells, dermal-derived), HMVEC-dBl-Neo (neonatal blood microvascular endothelial cells, dermal-derived), and HMVEC-dLy-Neo (neonatal lymphatic microvascular endothelial cells, dermal-derived). The relative DNaseI activity for each of these cell types is shown as black peaks. The location of the most significant SNP overall, rs4236601, overlapped with the transcription factor binding site c-Fos, highlighted in green.