Supplemental Material

Variant View: Visualizing Sequence Variants in their Gene Context.

Joel A. Ferstay, Cydney B. Nielsen, and Tamara Munzner

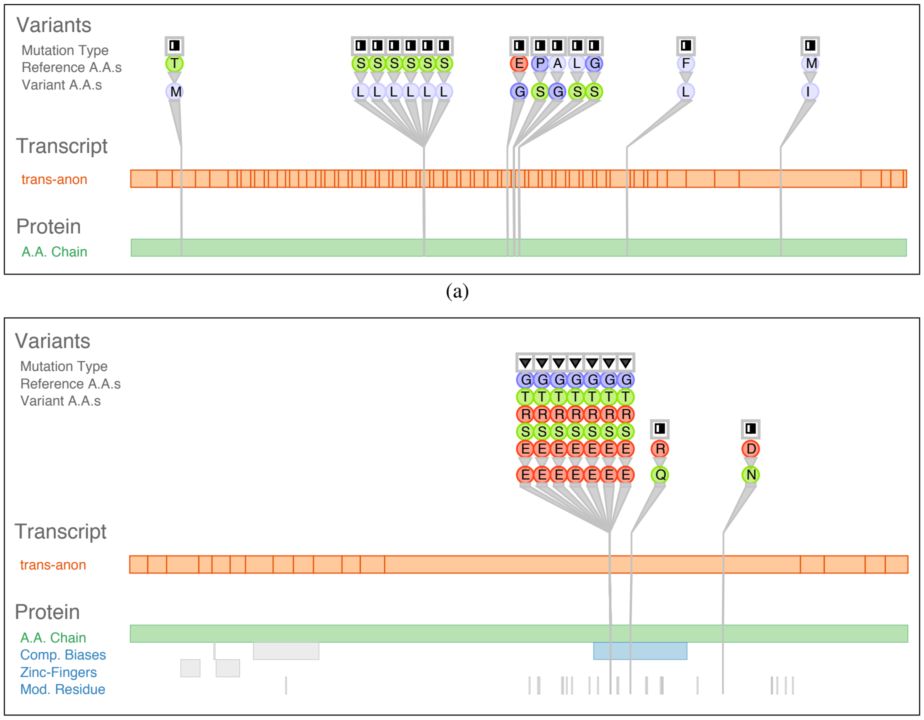
Abstract

We provide the following supplemental material for the paper "Variant View: Visualizing Sequence Variants in their Gene Context" by Joel A. Ferstay, Cydney B. Nielsen, and Tamara Munzner.

- Appendix A: Supplementary Figures
- Video: A demonstration of the Variant View tool. (QuickTime movie H.264 codec tested on Quicktime Version 10.1)

Appendix A: Supplementary Figures

In addition to retrieving and inspecting known variants in important AML genes, our analysts successfully used Variant View to discover interesting candidate genes. Figure S1(a) and (b) shows two more of these genes, in addition to the one shown in Figure 1 of the main paper. The gene names have been sanitized since their research is still ongoing and sample IDs in all examples have been sanitized to protect patient privacy. Figure S1(a) and (b) show either diverse or uniform variant types, respectively, within their hotspots that would not be detected by the hotspot metric alone. Interpretation of the biological importance of this variant diversity requires human judgement.



(a)

(b)

Figure 1: Variant View has proved useful for analysts in the discovery process of identifying new candidate genes. (Gene names sanitized as their follow-up research is still ongoing.) (a) The clear hotspots indicated a gene of interest. (b) The fact that the variants strike a range in a known function region type was the most informative aspect of this layout.

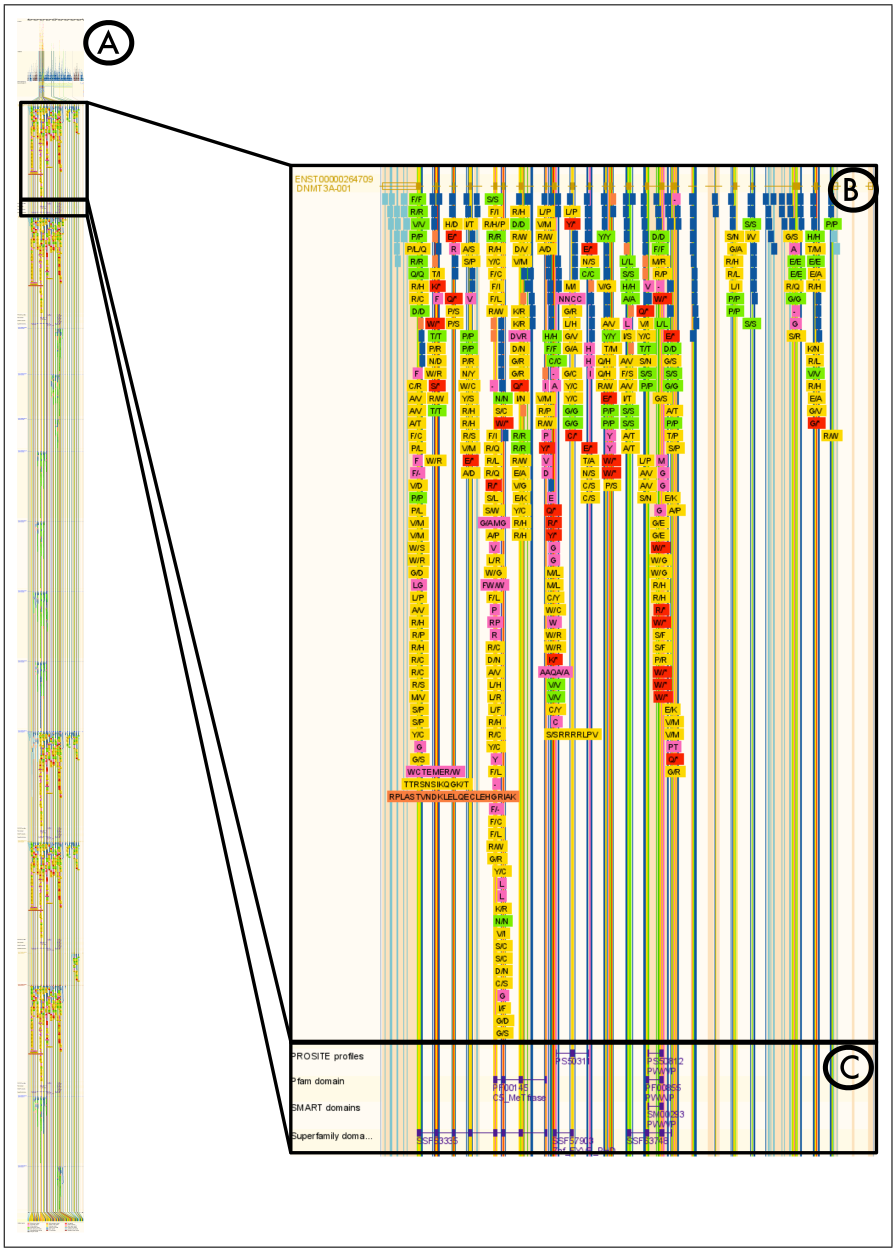


Figure 2: The Ensembl variation image's track-based view typically requires vertical scrolling, particularly to see variants across multiple alternative transcripts since each possible transcript and associated protein regions are stacked. The full display is labelled (A); this view extends across nearly seven pages when printed out directly from the browser. Variants are encoded as thin, vertical, colored lines. The region labelled (B) shows a magnified cropping of the display that includes a transcript scope similar to Variant View. The region labelled (C) shows protein regions which are also included in Variant View.