

The UCSC Genome Browser: new display modes to facilitate study of clinical data

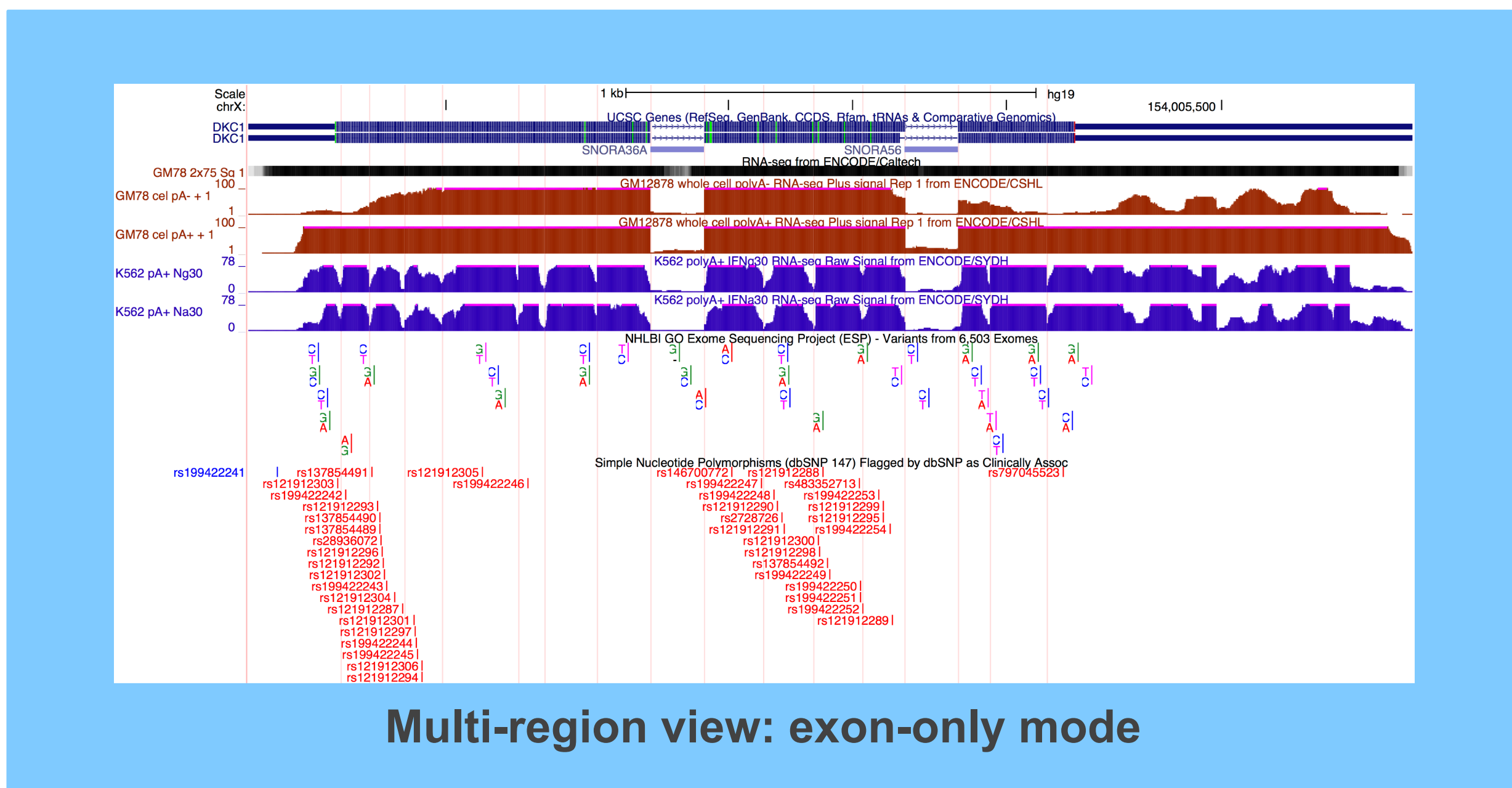
Matthew L. Speir¹, Galt P. Barber¹, Christopher M. Lee¹, Kate R. Rosenbloom¹, Angie S. Hinrichs¹, Cath Tyner¹, Luvina Guruvadoo¹, Brian T. Lee¹, Ann S. Zweig¹, Donna Karolchik¹, Robert M. Kuhn¹, W. James Kent¹
¹ University of California Santa Cruz Genomics Institute, Santa Cruz, CA, US

Introducing the new multi-region view

The UCSC Genome Browser (<https://genome.ucsc.edu/>) is a free, web-based tool that allows researchers to visualize and explore genomic data sets from sources including **OMIM**, **ClinVar**, and **GTEx** together with their own uploaded datasets.

One of the newest visualization tools in the UCSC Genome Browser is the multi-region configuration. This new tool allows you to display multiple, sometimes distant, regions of the genome alongside one another in the same Genome Browser image.

For example, using the new “**exon-only**” mode, we can visualize exome and dbSNP variants, and RNA sequencing results for an entire gene in greater detail:



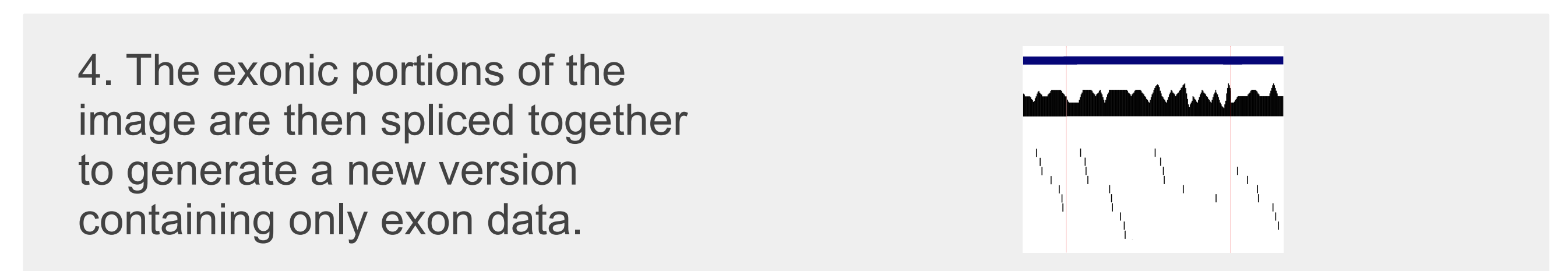
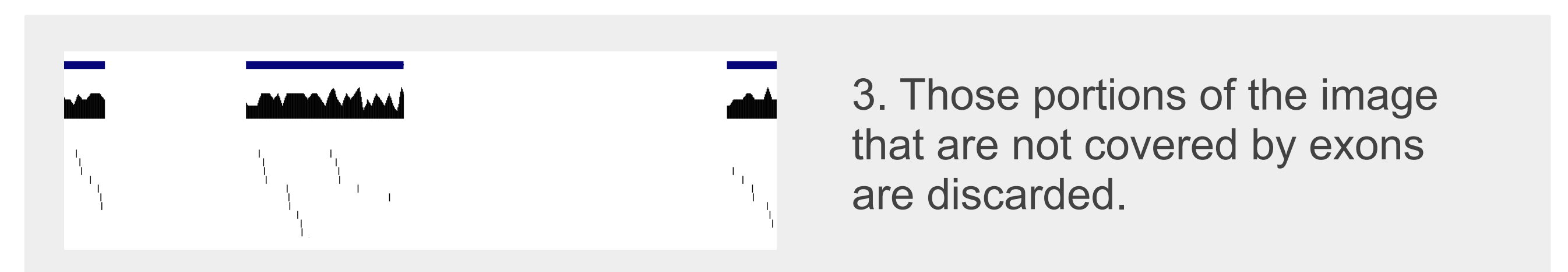
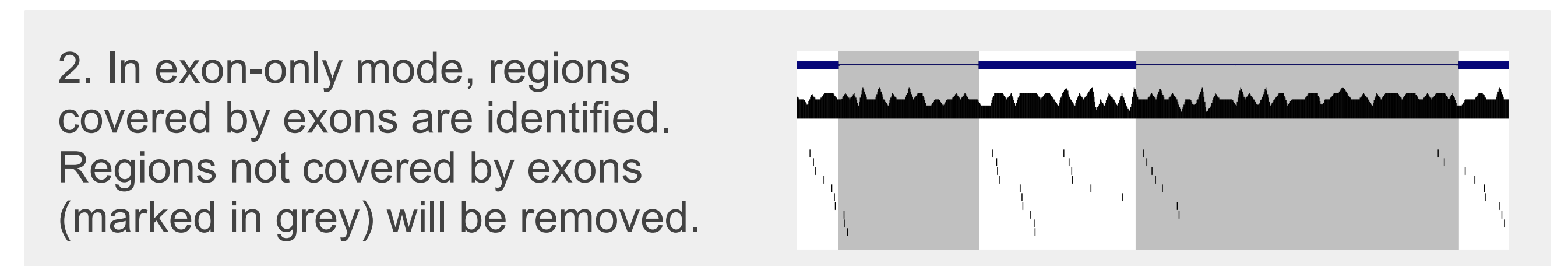
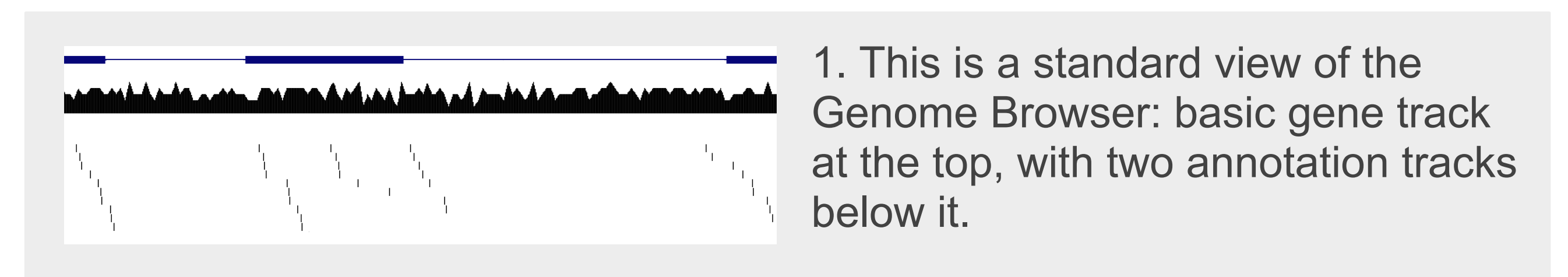
The new multi-region visualization modes can be accessed through:

- The **multi-region** button below the Genome Browser image
- The “View” menu above the Genome Browser image

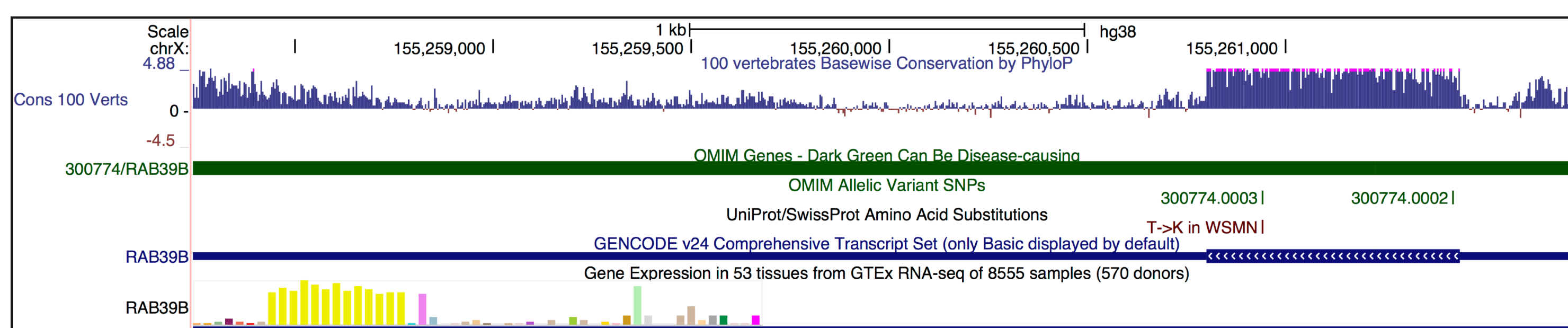
In addition to the **exon-only** mode demonstrated here, you can read about the other multi-region modes, such as **gene-only** or custom regions, in the following sections.

How does the multi-region exon-only view work?

In this breakdown, we will look at how the UCSC Genome Browser constructs the multi-region view for the **exon-only** mode. Labels and other details have been hidden for clarity and simplicity.

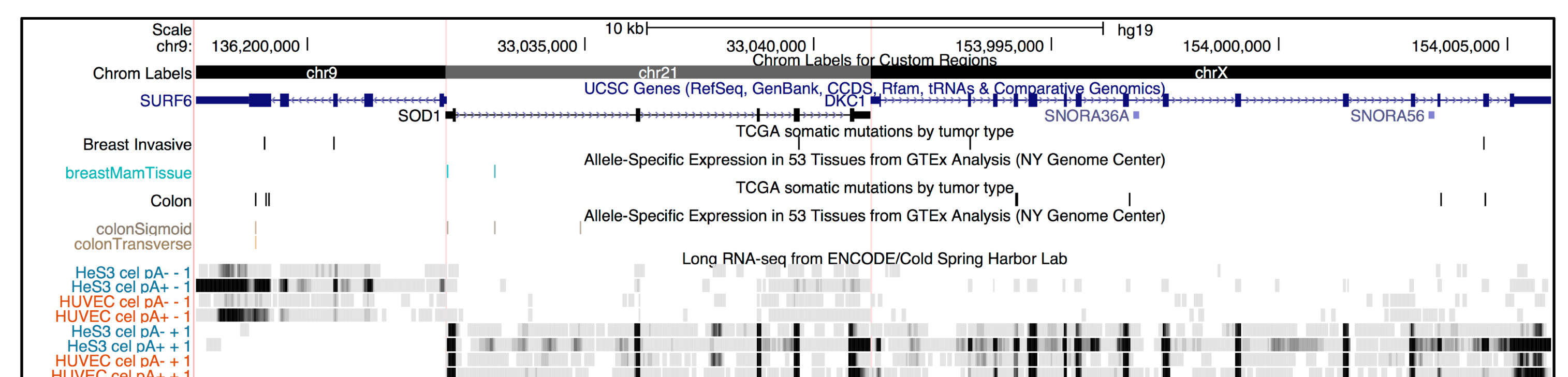
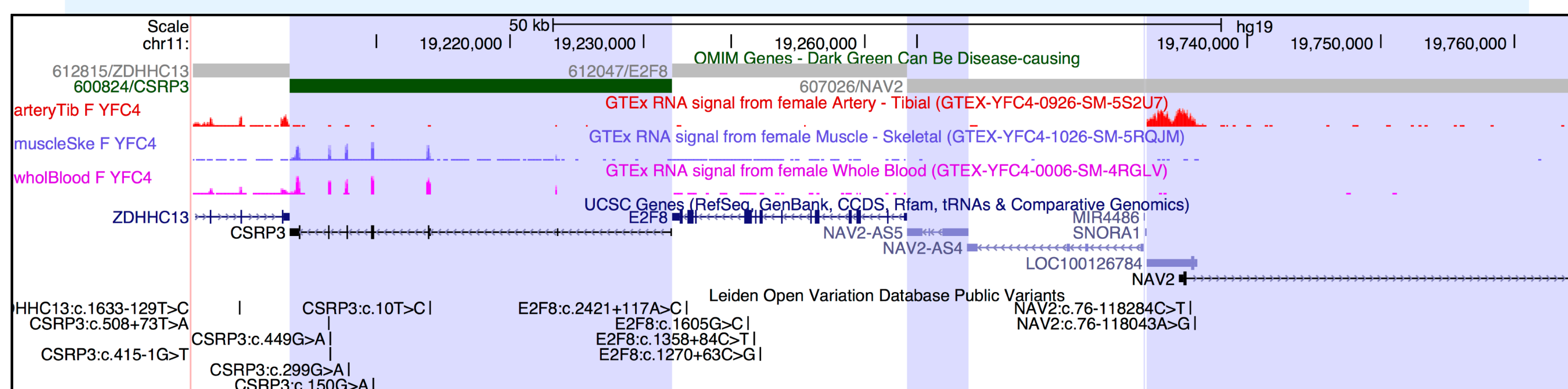


Current multi-region modes: exon-only, gene-only, custom regions, alternative haplotypes



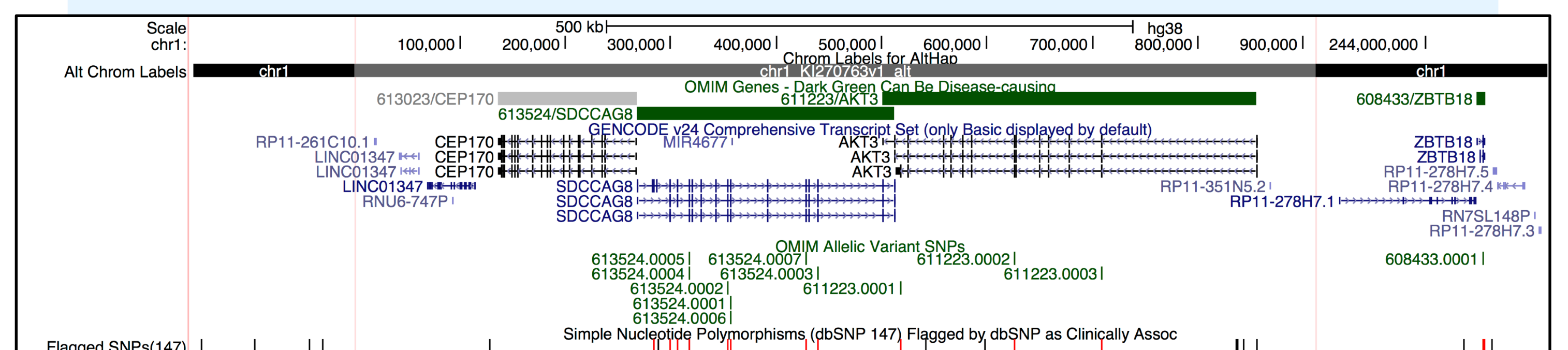
Exon-only: Allows you to remove the intronic and intergenic sequence from the display. Exonic regions are identified using a preselected gene annotation track and can be adjusted by user-defined padding bases. The display reacts to changes and filters applied to the gene set, removing regions no longer covered by exons or genes.

Gene-only: Allows you to remove only the intergenic regions from the Genome Browser display using a preselected gene annotation track. The gene regions are determined using the longest transcript in a region. If two genes overlap, then those are merged into a single gene region.



Custom regions: Allows you to display a set of regions that you define yourself. Regions do not have to be ordered or even on the same chromosome. Regions are uploaded using a plain text file made available on the web via HTTP.

Alternative haplotype: There are a number of alternative sequences provided with the current human reference genome. This mode inserts the alternative sequence and its annotations into the corresponding location on the reference genome, allowing you to view them within the greater context of the genome rather than as separate chromosomes.



Future work

- Allow for easier specification of custom regions:
 - Upload from a local file
 - Ability to paste-in regions
- Allow for the selection of different gene tracks for use with both **exon-only** and **gene-only** modes
- Allow assembly hubs to specify gene tracks for use with the **exon-only** and **gene-only** modes

More information

Multi-Region help:
genome.ucsc.edu/goldenPath/help/multiRegionHelp.html

Contact us: <https://genome.ucsc.edu/contacts.html>

Download this poster:
<http://genomewiki.ucsc.edu/index.php/Ashg2016Poster>

Follow us:
[@GenomeBrowser](https://twitter.com/GenomeBrowser) [/ucscGenomeBrowser](https://facebook.com/ucscGenomeBrowser) [UCSC Genome Browser](https://youtube.com/UCSCGenomeBrowser)

References

Speir ML, Zweig AS, Rosenbloom KR, Raney BJ, Paten B, Nejad P, Lee BT, Learned K, Karolchik D, Hinrichs AS, Heltner S, Harte RA, Haussler M, Guruvadoo L, Fujita PA, Eisenhart C, Diekhans M, Clawson H, Casper J, Barber GP, Haussler D, Kuhn RM, Kent WJ. The UCSC Genome Browser database: 2016 update. *Nucleic Acids Res.* 2016 Jan 4;44(D1):D717-25.

Funding and acknowledgements

Funding provided by National Human Genome Research Institute [5U41HG002371].

We would like to thank our loyal users, our collaborators and data contributors, and our tireless systems administrators for their support.

