Upgraded genome browsers at the Rat Genome Database support comparative and translational studies

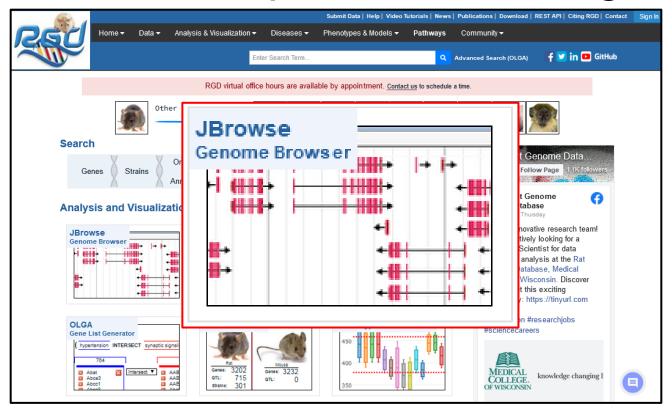
Jennifer R. Smith

20th Annual Meeting of the Complex Trait Community | Rat Genome & Models October 9, 2023





RGD is a multi-species knowledgebase



- Ten species: Human plus nine species used as models for human disease
- Standardized annotations for disease, qualitative and quantitative phenotypes, pathway, gene ontology, and gene-chemical interactions
- RGD has developed a suite of tools for finding and analyzing these data





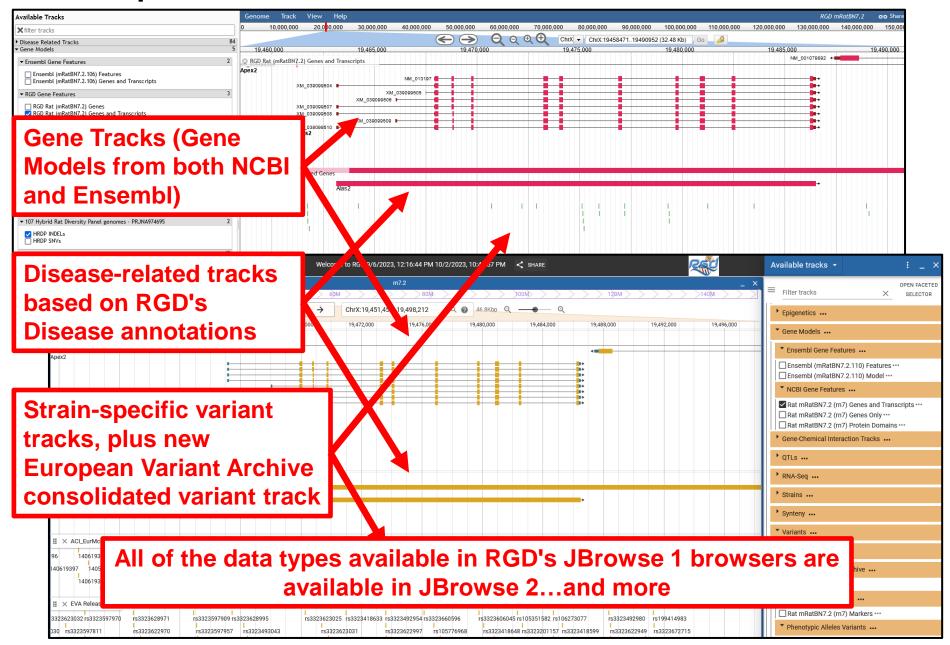
RGD is upgrading to JBrowse 2

- Currently released as a beta version
- Implemented for all RGD species
- Shares many features with other browsers
 - ➤ Search for the name or ID of a genome feature and go directly to that region
 - ➤ Select and view multiple data types in a single view
 - > Easy zooming and navigation across a chromosome
- Provides expanded functionality designed to facilitate comparative studies
- https://rgd.mcw.edu/jbrowse2/





Comparison of JBrowse 1 and JBrowse 2



New data types in JBrowse 2 Available tracks -OPEN FACETED Filter tracks Tracks ... Reference sequence (mRatBN7.2 (Epigenetics ••• Disease Related Tracks ... ATAC-Seq peaks and alignments for liver Epigenetics ••• ATAC-Seq ... and peaks for an additional 24 organs ATAC-Seq ... ChIP-Seq ... ChIP-Seg ··· ChIP-Seq peaks for methylation and Gene Models ••• acetylation marks and TF binding sites in * Ensembl Gene Features ••• liver and denervated and non-denervated Ensembl (mRatBN7.2.110) Features *** Ensembl (mRatBN7.2.110) Model *** gastrocnemius muscle NCBI Gene Features ... Rat mRatBN7.2 (rn7) Genes and Transcripts *** Rat mRatBN7.2 (rn7) Genes Only *** Synteny ... Rat mRatBN7.2 (rn7) Protein Domains *** Gene-Chemical Interaction Tracks ••• rn7.2 to canFam3 Synteny *** OTLs ... rn7.2 to hg38 Synteny *** RNA-Seq ... rn7.2 to mm39 Synteny *** Strains ... rn7.2 to rn3.4 Synteny *** Synteny ... rn7.2 to rn5 Synteny *** ▼ Variants ••• rn7.2 to rn6 Synteny *** Damaging Variants ... rn7.2 to susScr11 Synteny *** European Variation Archive ••• rn7.2 to UTH_Rnor_SHR_Utx Synteny ••• Micro Satellite Markers ••• rn7.2 to UTH_Rnor_SHRSP_BbbUtx_1.0 Synteny *** Phenotypic Alleles Variants ... rn7.2 to UTH_Rnor_WKY_Bbb_1.0 Synteny *** Strain Specific Variants ...

New data types in JBrowse 2

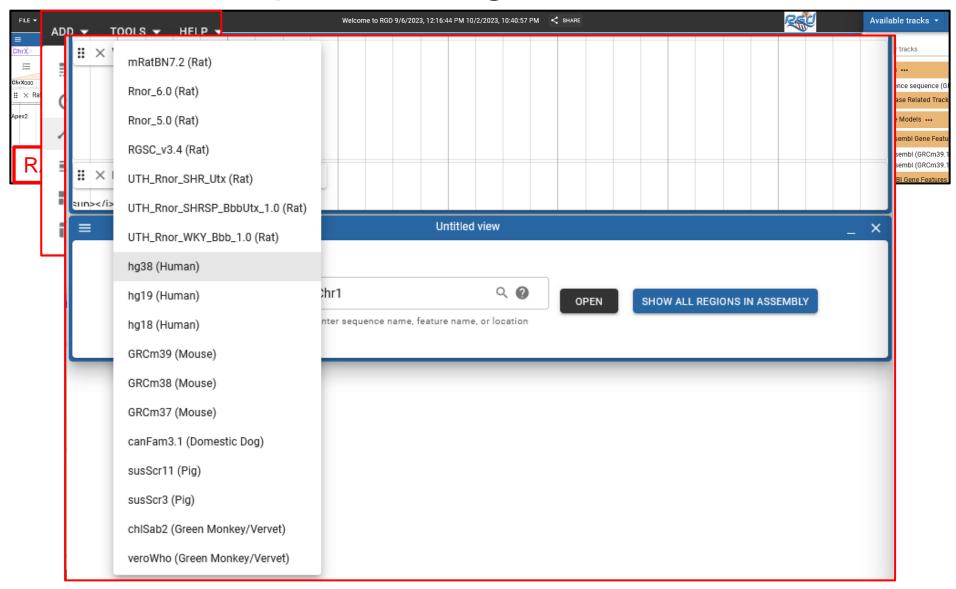


The right sidebar can display either a track selector or feature details, in this case, it shows information about the Mthfr gene. The sidebar is on the right by default but can be moved to the left of the main genome browser subwindow.





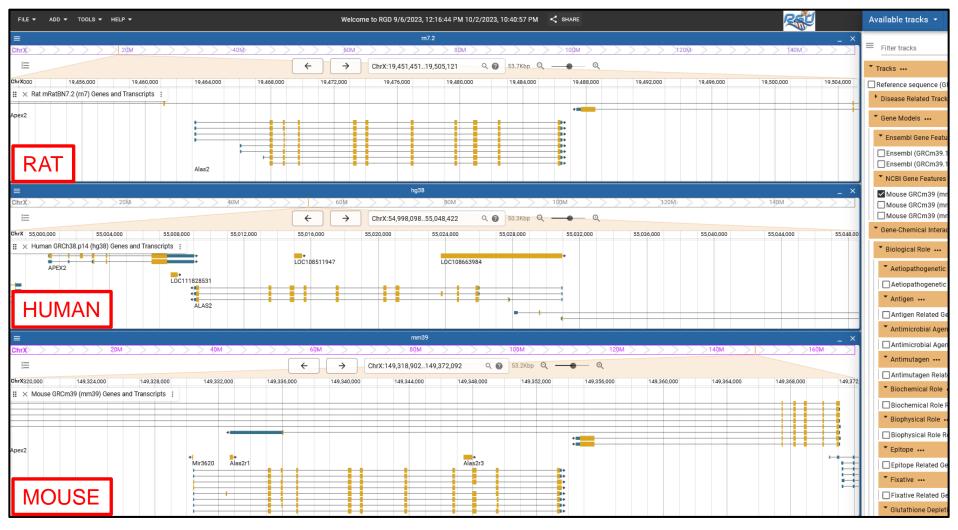
Multiple linear genome views







Multiple linear genome views

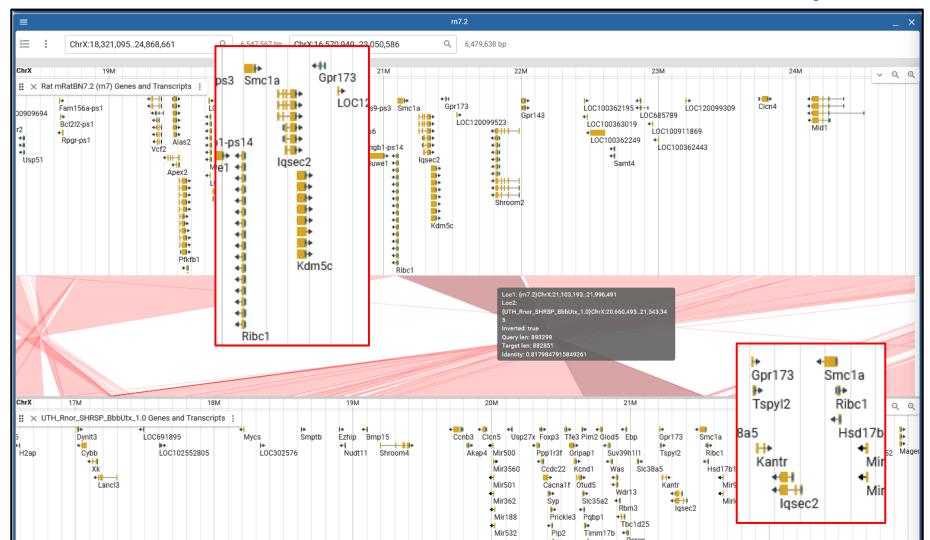


The stacked linear genome views make it easy to see that in mouse and rat, Alas2 overlaps Apex2 whereas in human ALAS2 is downstream of APEX2.





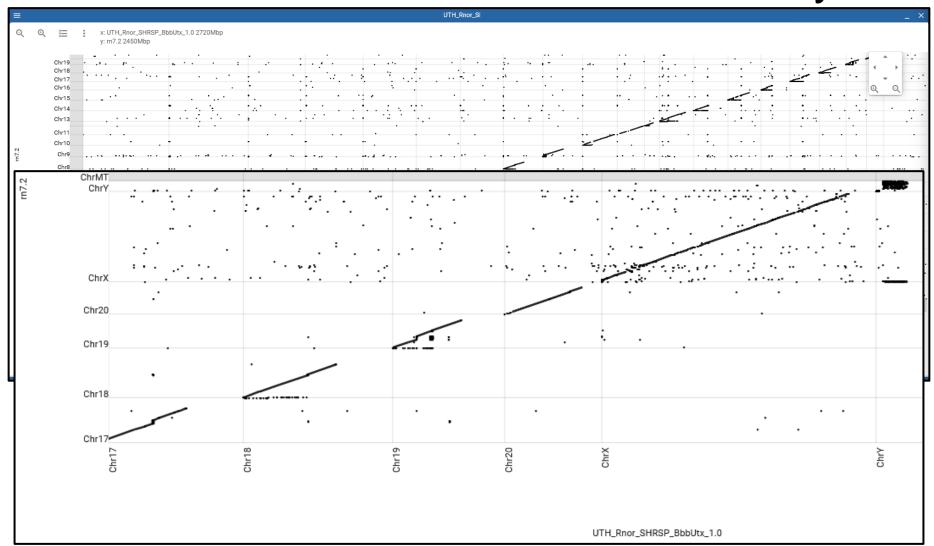
Synteny view: mRatBN7.2 vs. SHRSP/BbbUtx *de novo* assembly







Dotplot View: mRatBN7.2 vs. SHRSP/BbbUtx *de novo* assembly

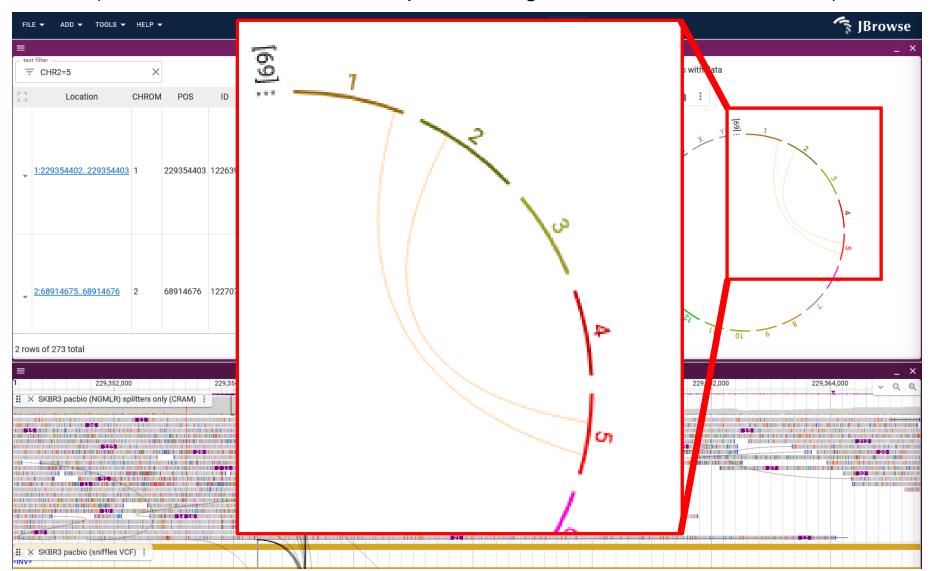






Breakpoint Split View and Circular Plot

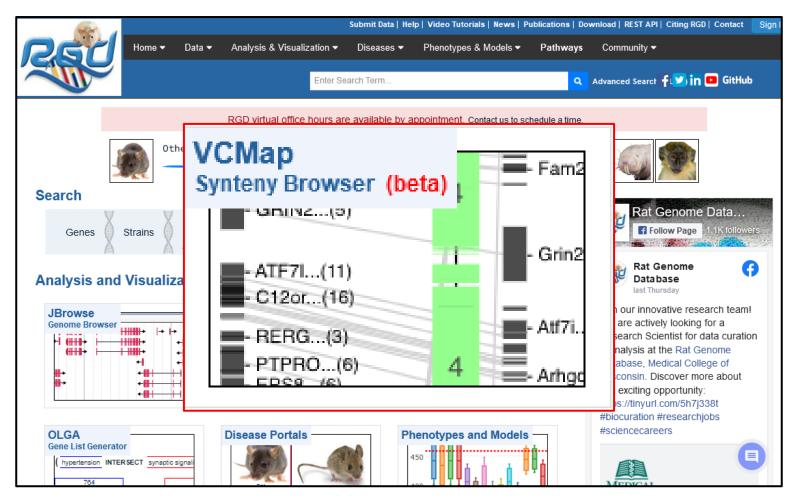
(aka the Structural Variant inspector, image from the JBrowse website)







Viewing synteny using VCMap

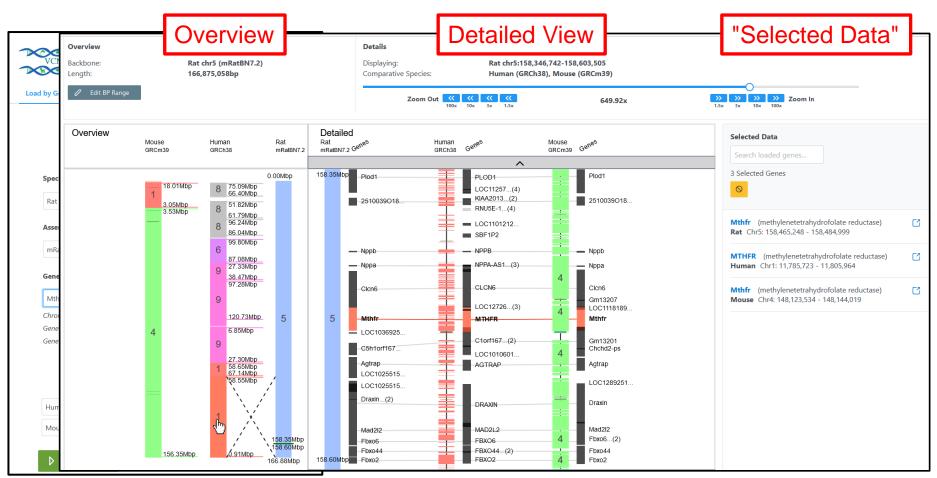


VCMap is a synteny viewer that allows comparisons across multiple species or assemblies.





Viewing synteny using VCMap

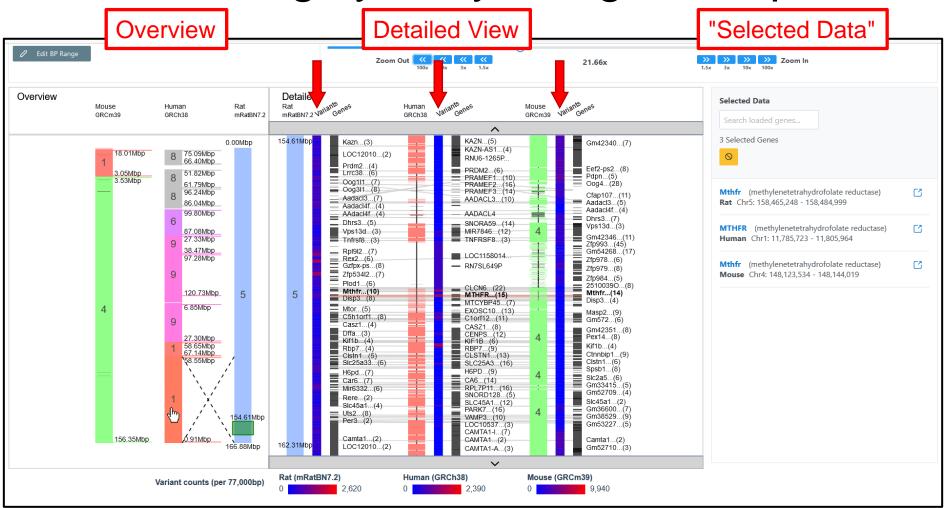


The overview panel gives a chromosome-wide view of the syntenic blocks. The detailed view shows blocks and gaps with a genes track for each species. When a gene or gene group is selected, the orthology is indicated by a red line and information is displayed in the "Selected Data" panel with links to gene records in RGD.





Viewing synteny using VCMap



Heatmap tracks for variant density in the three species are currently available. Variants are binned, with bin sizes dependent on the zoom level and colors determined by the number of variants in the bin. Heatmaps for epigenome data are coming soon.





The RGD Team:

Principal Investigator:
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Co-Investigator:

Mindy Dwinell, PhD

Curation Team:

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Database Administrator: Stacy Zacher, MSc

Thank you!

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(grant 2P01HL082798 from the National Heart, Lung, and Blood Institute),

Dr. Peter Doris for the strain-specific *de novo* assembly data, the members of the International Rat Omics Consortium (IROC), **And to the researchers** who contribute data and who faithfully use our website and data!

JBrowse and JBrowse 2 were developed by The JBrowse Consortium (https://jbrowse.org/; https://github.com/GMOD/jbrowse-components)











GLOBAL CORE BIODATA RESOURCE



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