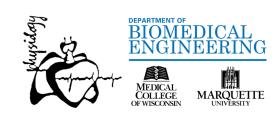
The Rat Genome Database resources for de novo rat strain assemblies

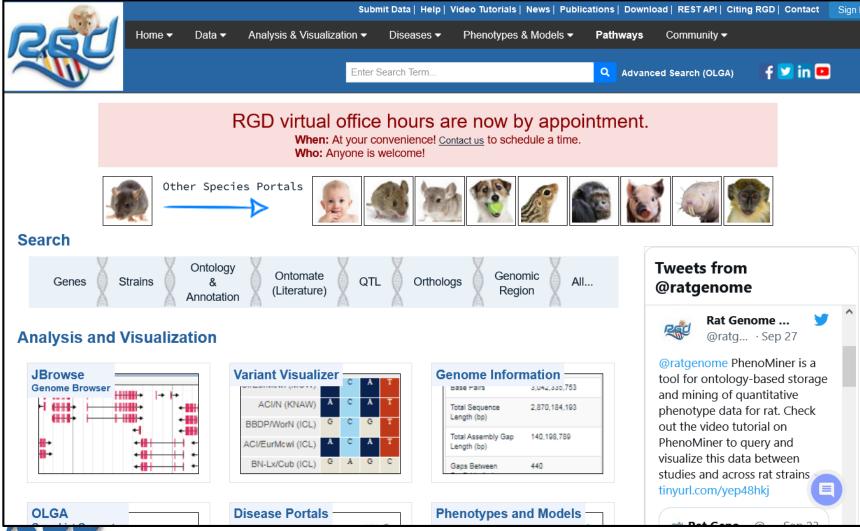
Jennifer R. Smith

Complex Trait Community and Rat Genomics meeting September 29, 2022





RGD is a cross-species knowledgebase providing data linking disease to the genome



RGD is committed to the integration of the full spectrum of genomic data to support research into the underlying causes of human disease.





WGS and de novo assembly resulted in reference-quality assemblies for strains SHR/Utx, SHRSP/BbbUtx and WKY/Bbb

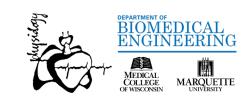
- The SHR and SHRSP strains are models of hypertension and stroke; WKY is a commonly used control strain for hypertension
- The sequencing, performed by Dr. Peter Doris and colleagues, employed long-read technologies (PacBio, HiC and BioNano)
- Long reads facilitated the assembly of highly repetitive regions, e.g., the Y chromosome
 - mRatBN7.2 Y has an assembled length of 18,315,841
 - ➤ UTH_Rnor_SHR_Utx Y has an assembled length of 32,175,099
 - UTH_Rnor_SHRSP_BbbUtx_1.0 Y has an assembled length of 38,271,881
 - ➤ UTH_Rnor_WKY_Bbb_1.0 Y has an assembled length of 31,627,559
- NCBI RefSeq did genome annotation to map genes and transcripts onto the new assemblies but have not performed manual curation



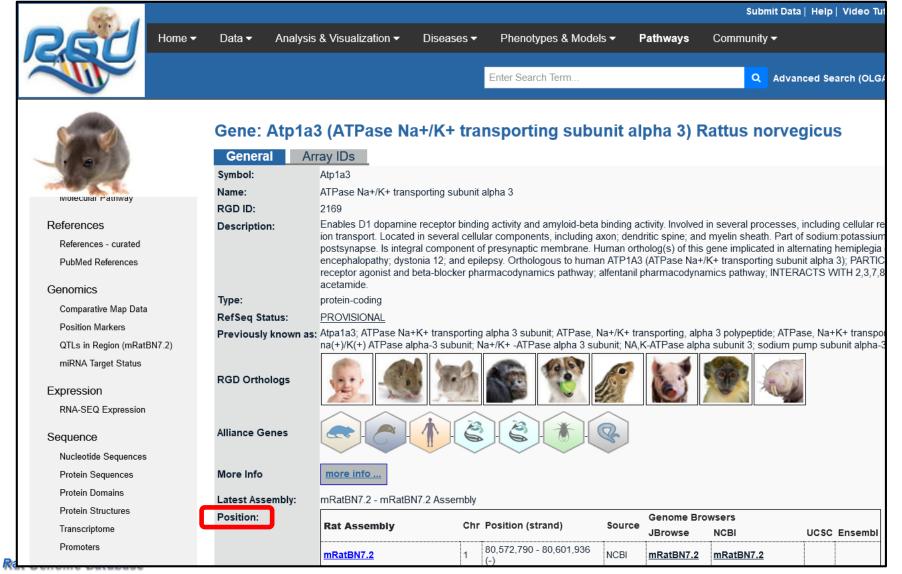
Data for the SHR, SHRSP and WKY strains are available on RGD

• For genes that have been mapped to any or all of these assemblies, positions are available on the gene report pages.





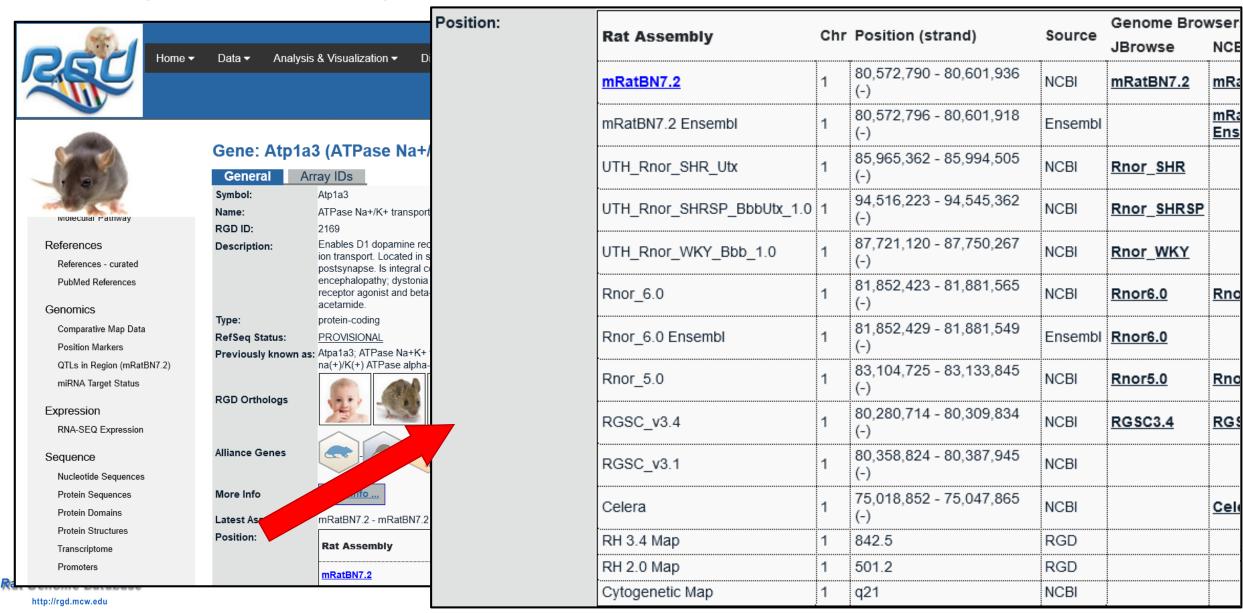
RGD maintains positions for all assemblies and maps on gene pages, including the SHR, SHRSP and WKY assemblies







RGD maintains positions for all assemblies and maps on gene pages, including the SHR, SHRSP and WKY assemblies



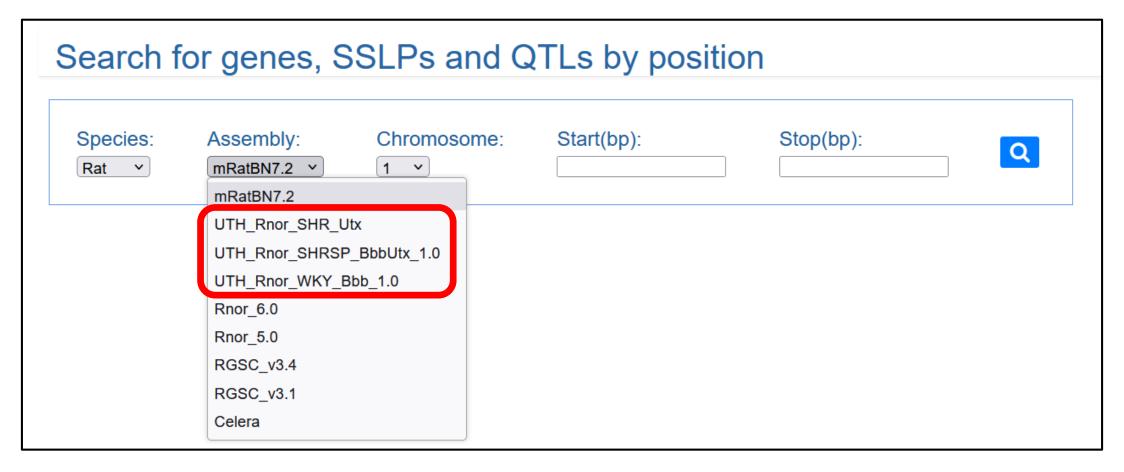
Data for the SHR, SHRSP and WKY strains are available on RGD

- For genes that have been mapped to any or all of these assemblies, positions are available on the gene report pages.
- The SHR, SHRSP and WKY assemblies are included in RGD's Search by Position tool





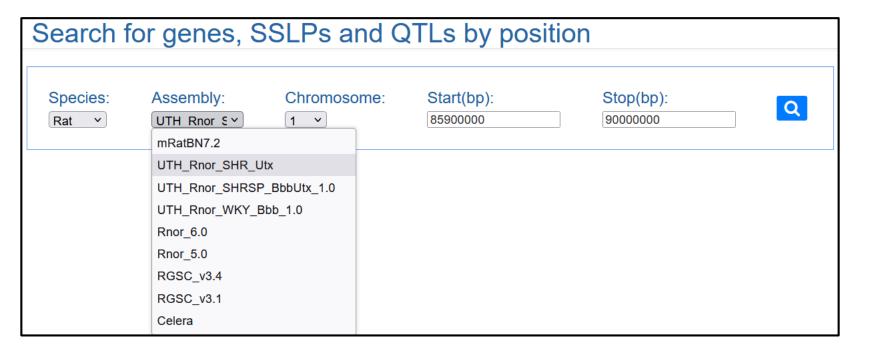
Search by position is available for the new assemblies



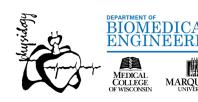




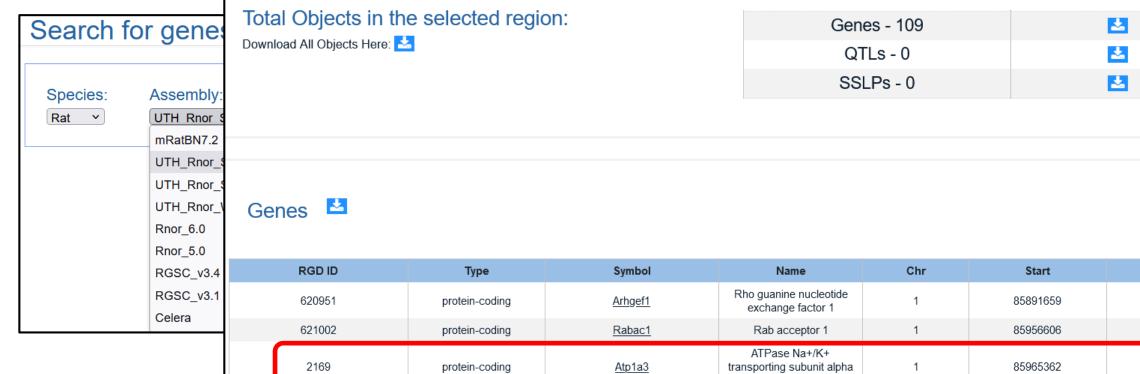
Search by position is available for the new assemblies







Search by position is available for the new assemblies





621002	protein-coding	Rabac1	Rab acceptor 1	1	85956606	85959730
2169	protein-coding	Atp1a3	ATPase Na+/K+ transporting subunit alpha 3	1	85965362	85994505
2735	protein-coding	<u>Grik5</u>	glutamate ionotropic receptor kainate type subunit 5	1	85998459	86059573
1311420	protein-coding	<u>Zfp574</u>	zinc finger protein 574	1	86064885	86070148
621690	protein-coding	Pou2f2	POU class 2 homeobox 2	1	86079615	86116737
620351	protein-coding	<u>Gsk3a</u>	glycogen synthase kinase 3 alpha	1	86208062	86217908
1311991	protein-coding	<u>Erf</u>	Ets2 repressor factor	1	86222152	86230605
1310706	protein-coding	<u>Cic</u>	capicua transcriptional	1	86246136	86272754

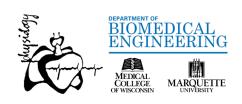
Stop

85913491

Data for the SHR, SHRSP and WKY strains are available on RGD

- For genes that have been mapped to any or all of these assemblies, positions are available on the gene report pages.
- The SHR, SHRSP and WKY assemblies are included in RGD's Search by Position tool
- RGD has created JBrowse genome browsers for the SHR, SHRSP and WKY assemblies

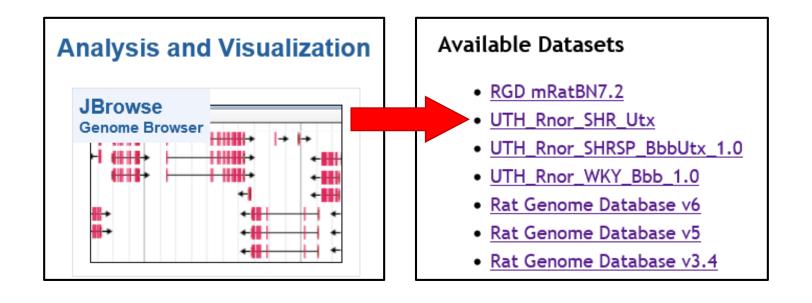




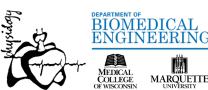
RGD has created JBrowse genome browsers for the SHR, SHRSP and WKY assemblies

Position:	Rat Assembly	Chr Position (strand)		Source	Genome Brow JBrowse
	mRatBN7.2	1	80,572,790 - 80,601,936 (-)	NCBI	mRatBN7.2
	mRatBN7.2 Ensembl	1	80,572,796 - 80,601,918 (-)	Ensemi	
	UTH_Rnor_SHR_Utx	1	85,965,362 - 85,994,505 (-)	NCBI	Rnor_SHR
	UTH_Rnor_SHRSP_BbbUtx_1.0		94,516,223 - 94,545,362 (-)	NCBI	Rnor_SHRSP
	UTH_Rnor_WKY_Bbb_1.0	1	87,721,120 - 87,750,267 (-)	NCBI	Rnor_WKY

Access JBrowse from the position section of the gene page, or use the link in the Analysis and Visualization section of the RGD home page.





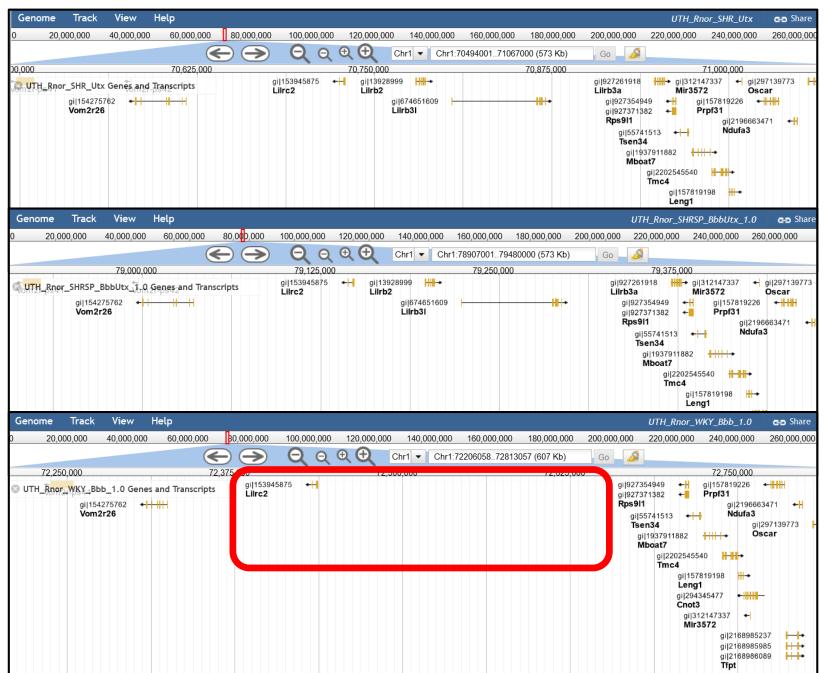




SHR

SHRSP





Looking at the region around the gene Lilrc2 you can see that SHR and SHRSP have two genes, Lilrb2 and Lilrb3l, that are missing from the WKY assembly







Data for the SHR, SHRSP and WKY strains are available on RGD

- For genes that have been mapped to any or all of these assemblies, positions are available on the gene report pages.
- The SHR, SHRSP and WKY assemblies are included in RGD's Search by Position tool
- RGD has created JBrowse genome browsers for the SHR, SHRSP and WKY assemblies
- RGD has produced pairwise gene comparison files for all combinations of mRatBN7.2, SHR, SHRSP and WKY



RGD has produced pairwise gene position comparison files

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Index of /data_release Name	Last modified Size	Each folder contains files for genes that map to different chromosomes in the two assemblies and the							
GENES_BONOBO.txt	2022-09-23 23:41 17M	genes that are only found on one or							
GENES_CHINCHILLA.txt	2022-09-23 23:41 14M	the other assembly.							
GENES_DOG.txt	2022-09-23 23:42 35M								
GENES_HUMAN.txt	Name	Date modified Type							
GENES_MOUSE.txt	nn7 vs 3.4	9/27/2022 11:41 AM File folder							
GENES_OBSOLETE_IDS.txt	nn7 vs rn5	9/27/2022 11:41 AM File folder							
GENES_PIG.txt GENES_RAT.txt	📜 rn7 vs rn6	9/27/2022 11:41 AM File folder							
GENES_RAT_assembly_comparison/	UTH_Shr vs rn7	9/27/2022 11:41 AM File folder							
GENES_README	UTH_Shr vs UTH_Shi	rsp 9/27/2022 11:41 AM File folder							
<u> </u>	UTH_Shr vs UTH_Wk	Y 9/27/2022 11:41 AM File folder							
	UTH_Shrsp vs rn7	9/27/2022 11:41 AM File folder							
	UTH_Shrsp vs UTH_\	WKY 9/27/2022 11:41 AM File folder							

UTH WKY vs rn7

9/27/2022 11:41 AM

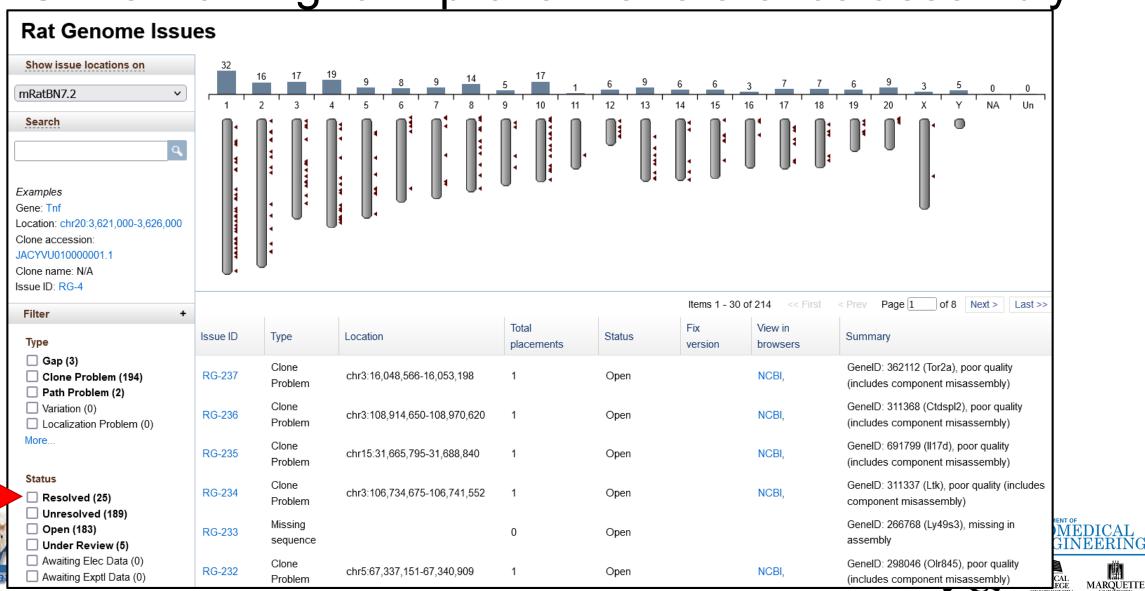
File folder



As a member of the Genome Reference Consortium, RGD is working to improve the reference assembly

- NCBI's RefSeq curators reported 214 genomic regions with suspected assembly problems.
- Dr. Peter Doris also created a list of 287 problematic genes from analysis of the mRatBN7.2 assembly compared to the SHR, SHRSP and WKY assemblies.
- Of those 287 genes, 179 were also in the group reported by RefSeq curators.
- RGD curator Wendy Demos has closed 25 tickets, 24 of which are from the overlap group

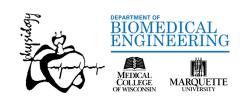
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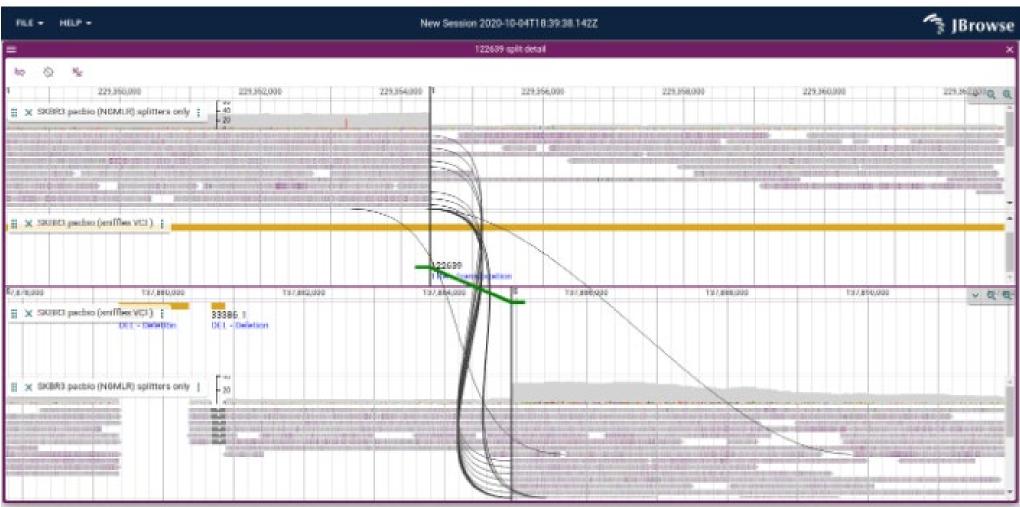
RGD is upgrading our genome browsers to JBrowse 2.0

- JBrowse 2 is more feature-rich and has functionality that is designed to facilitate comparisons, including...
 - ➤ Breakpoint Split Viewer
 - ➤ Whole Genome Alignment Dotplot Viewer
 - ➤ Linear Synteny Viewer





Breakpoint Split Viewer

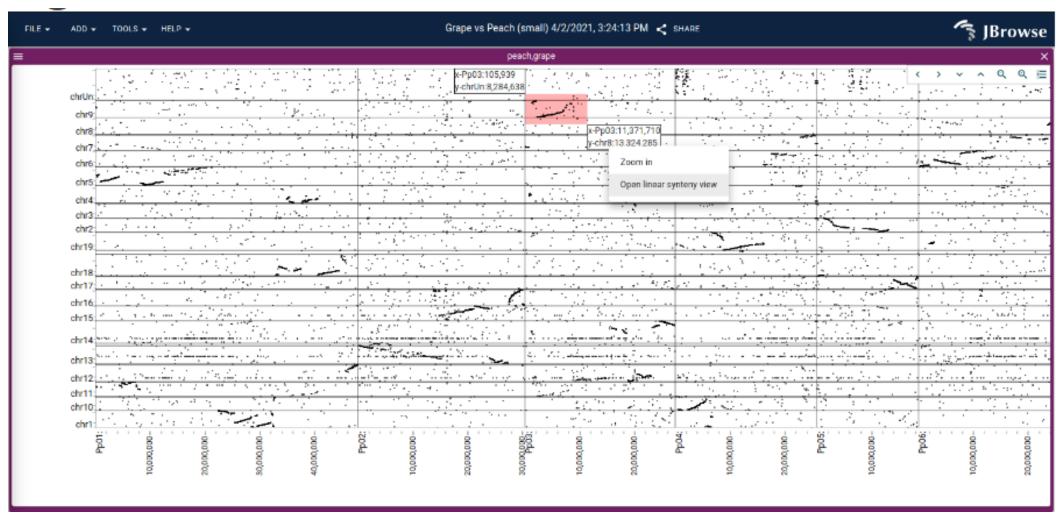








Whole Genome Alignment Dotplot Viewer

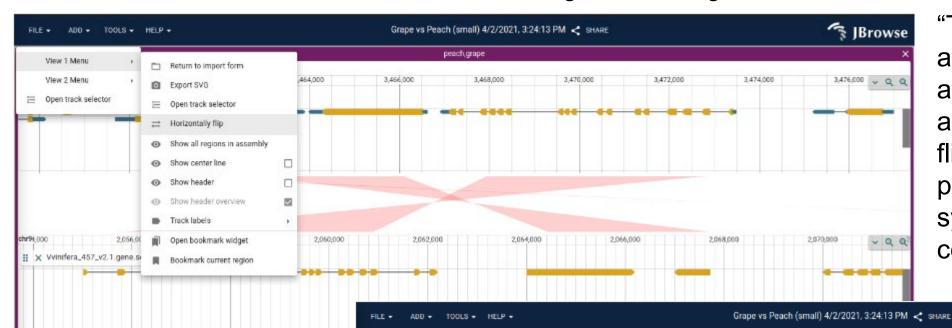








Linear Synteny Viewer



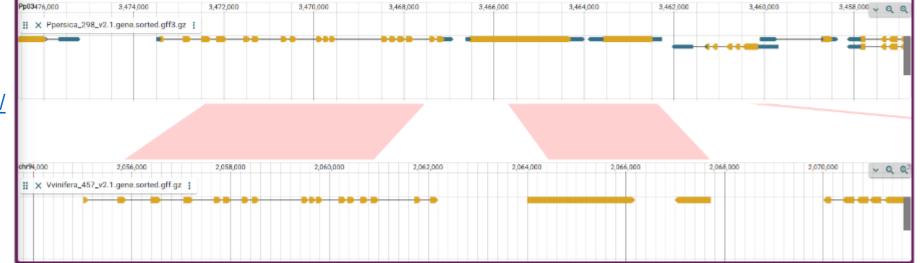
80 10 %

"The [top] picture shows an inverted region being aligned, and the [bottom] applies the 'horizontally flip' action to the top panel, allowing the synteny to be viewed in a co-linear way"

IBrowse







peach,grape

The RGD Team:

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Thank you!



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Our collaborators, Dr. Peter Doris and the IROC consortium, and the researchers who contribute data and who faithfully use our website and data!

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