## Expansion of Rat Expression Data in the Rat Genome Database

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20th annual Complex Trait Community | Rat Genomics Community meeting, 8-12 October 2023







Gene: Gjb2 (gap junction protein, beta 2) Rattus norvegicus

General	Aı
Symbol:	
Name:	

#### Summary

#### Annotation

RGD Manual Disease Imported Disease - ClinVar Imported Disease - CTD Imported Disease - MGI Imported Disease - OMIM Gene-Chemical Interaction

Gene Ontology

#### References

References - curated PubMed References

#### Genomics Variants

Comparative Map Data

miRNA Target Status

QTLs in Region (mRatBN7.2)

Markers in Region

#### Expression RNA-SEQ Expression

#### Sequence

Nucleotide Sequences

Reference Sequences

Protein Sequences

Reference Protein Sequences

Protein Domains Protein Structures

Transcriptome

#### Additional Information

External Database Links

Nomenclature History **RGD Curation Notes** 

Gjb2

gap junction protein, beta 2

RGD ID: Description:

Enables gap junction channel activity. Involved in several processes, including cellular response to glucagon stimulus; decidualization; and re projection; gap junction; and lateral plasma membrane. Used to study hepatocellular carcinoma and lung adenocarcinoma. Biomarker of aut urethral obstruction. Human ortholog(s) of this gene implicated in Bart-Pumphrey syndrome; Vohwinkel syndrome; autosomal dominant kera keratoderma-deafness syndrome. Orthologous to human GJB2 (gap junction protein beta 2); INTERACTS WITH 17alpha-ethynylestradiol; 1

protein-coding

VALIDATED RefSeq Status:

Previously known as: connexin 26; connexin e; connexin

**RGD Orthologs** 

















31.27 Full-screen view

**Alliance Genes** 



Latest Assembly:

mDatDN7.2 mDatDN7.2 Assamble

Position:

JBrowse:

More Info

Rat Assembly		Position (strand)	Source	Genome Browsers				
		Position (straint)		JBrowse	NCBI	UCSC	Ensembl	
mRatBN7.2	15	31,260,390 - 31,278,222 (-)	NCBI	mRatBN7.2	mRatBN7.2			
mRatBN7.2 Ensembl	15	31,260,357 - 31,278,177 (-)	Ensembl		mRatBN7.2 Ensembl			
UTH_Rnor_SHR_Utx	15	33,251,720 - 33,257,696 (-)	NCBI	Rnor SHR	UTH Rnor SHR Utx			
UTH_Rnor_SHRSP_BbbUtx_1.0	15	34,402,373 - 34,408,349 (-)	NCBI	Rnor SHRSP	UTH Rnor SHRSP BbbUtx 1.0			
UTH_Rnor_WKY_Bbb_1.0	15	32,660,503 - 32,666,480 (-)	NCBI	Rnor WKY	UTH Rnor WKY Bbb 1.0			
Rnor_6.0	15	37,377,313 - 37,394,494 (-)	NCBI	Rnor6.0	Rnor 6.0	<u>rn6</u>	Rnor6.0	
Rnor_6.0 Ensembl	15	37,377,316 - 37,383,277 (-)	Ensembl	Rnor6.0		<u>rn6</u>	Rnor6.0	
Rnor_5.0	15	41,224,497 - 41,236,203 (-)	NCBI	Rnor5.0	Rnor 5.0	<u>rn5</u>	Rnor5.0	
RGSC_v3.4	15	36,153,526 - 36,159,490 (-)	NCBI	RGSC3.4	RGSC v3.4	<u>rn4</u>	RGSC3.4	
RGSC_v3.1	15	36,169,226 - 36,175,190 (-)	NCBI					
Celera	15	30,972,813 - 30,978,777 (-)	NCBI		<u>Celera</u>			
Cytogenetic Map	15	p12	NCBI					





XM\_039093557 ←■

RGD Rat (mRatBN7.2) Genes and Transcripts

#### RNA-SEQ Expression

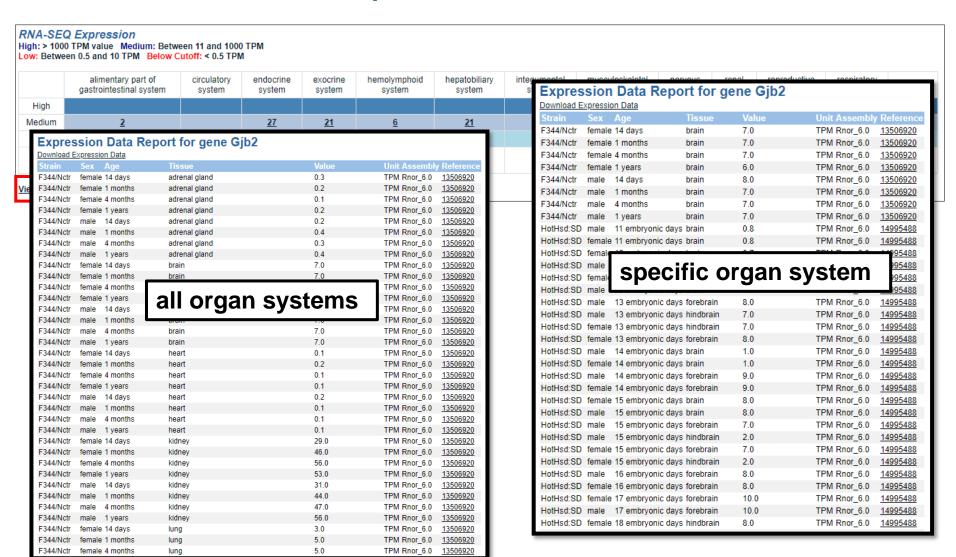
High: > 1000 TPM value Medium: Between 11 and 1000 TPM Low: Between 0.5 and 10 TPM Below Cutoff: < 0.5 TPM

	alimentary part of gastrointestinal system	circulatory system	endocrine system	exocrine system	hemolymphoid system	hepatobiliary system	integumental system	musculoskeletal system	nervous system	renal system	reproductive system	respiratory system	appendage
High													
Medium	<u>2</u>		<u>27</u>	<u>21</u>	<u>6</u>	<u>21</u>			<u>34</u>	<u>23</u>	<u>3</u>		
Low	1	<u>20</u>	<u>22</u>	<u>20</u>	<u>13</u>	<u>20</u>	<u>3</u>	<u>4</u>	<u>38</u>	<u>12</u>	<u>27</u>	<u>11</u>	<u>3</u>
Below cutoff		<u>21</u>	8				<u>5</u>	7	<u>2</u>		<u>11</u>		<u>5</u>

View RNA-SEQ Expression Data











Species	Study Source	# Studies	# Records
Human	EXPR-ATLAS	22	20,972
Rat	EXPR-ATLAS	3	313
Rat	GEO	4	448
Dog	GEO	1	75
Pig	EXPR-ATLAS	1	51
Vervet	EXPR-ATLAS	1	116

31 studies

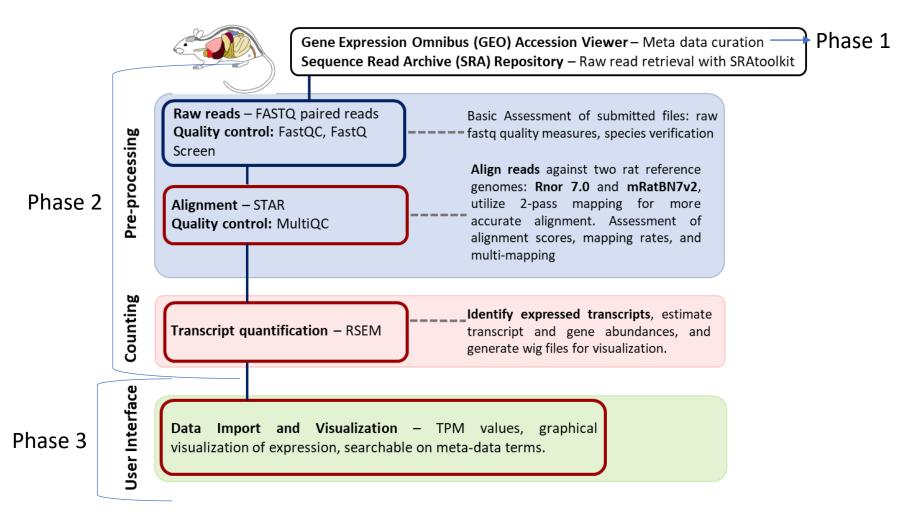
21,975 records

Approximately 32,000 individual expression values per record





### Data Re-mapping



Dobin A, Davis C, Schlesinger F, Drenkow J, Zaleski C, Jha S, Batut P, Chaisson M, Gingeras T: STAR: ultrafast universal RNA-seq aligner. Bioinformatics 2012, Jan:29(1): 15-21

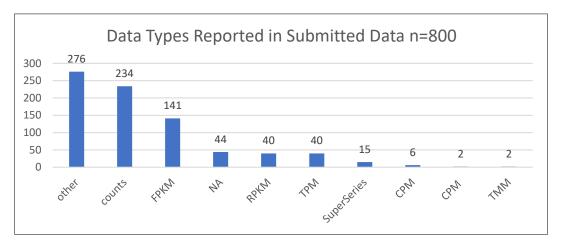
Li B, Dewey C: RSEM: accurate transcript quantification from RNA-Seq data with or without a reference genome. BMC Bioinformatics 2011 (12), 323





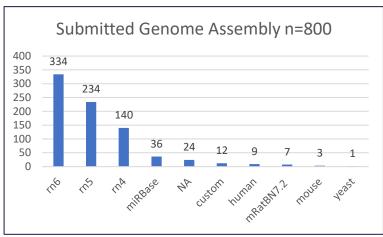
#### **GEO Rat Expression Accession Assessments**

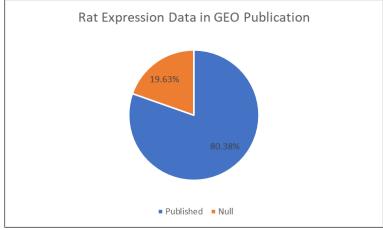
- 1,135 Rat Expression Studies imported to RGD Expression Curation Tool
- 800 studies reviewed



Other: Differential expression, methylation, UMI, etc. SuperSeries: multiple data types are often provided

NA: unknown, conflicts from what is stated as submitted data type

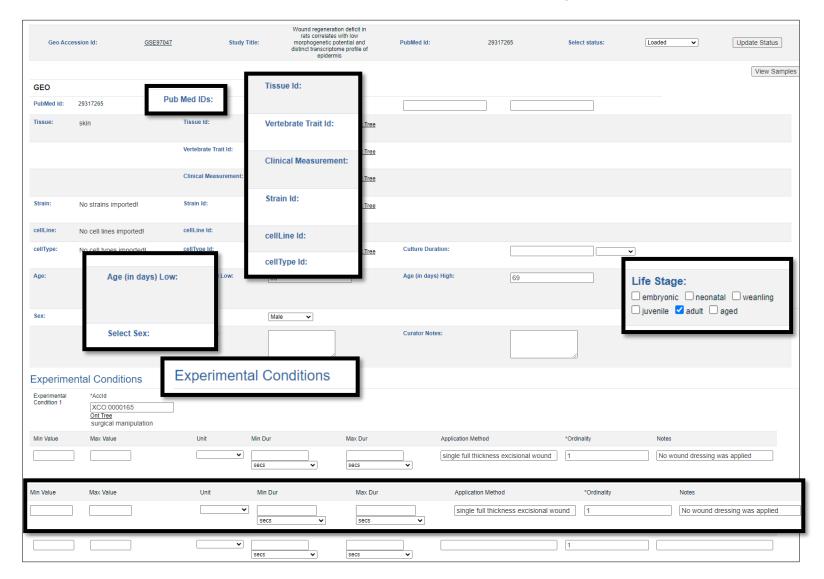








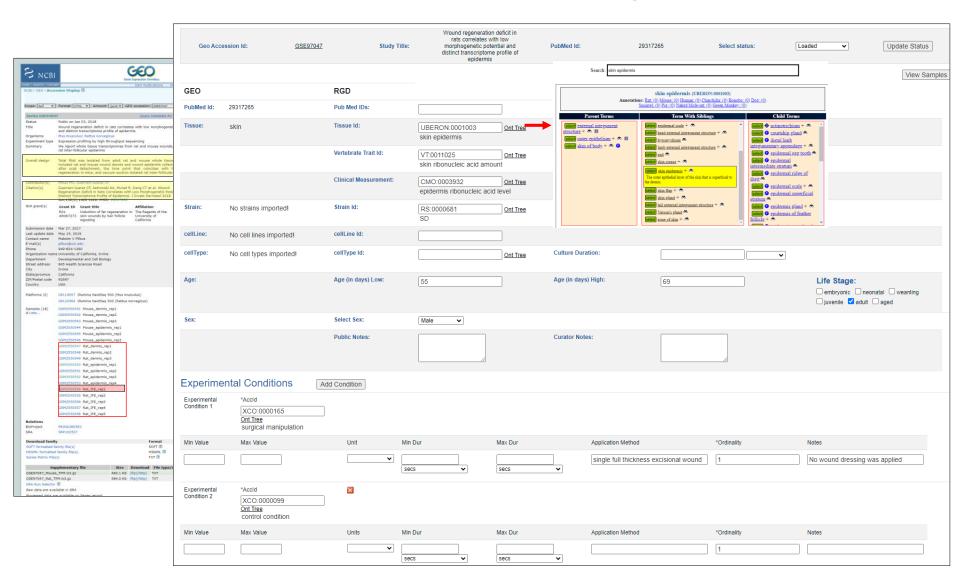
## Metadata Curation - Project Level







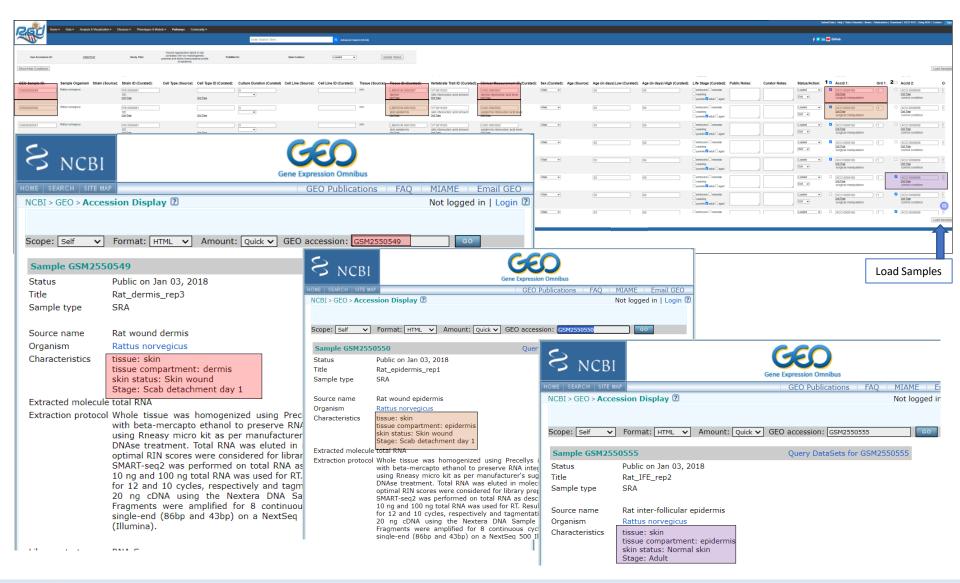
# Metadata Curation - Project Level







## Sample Level Curation







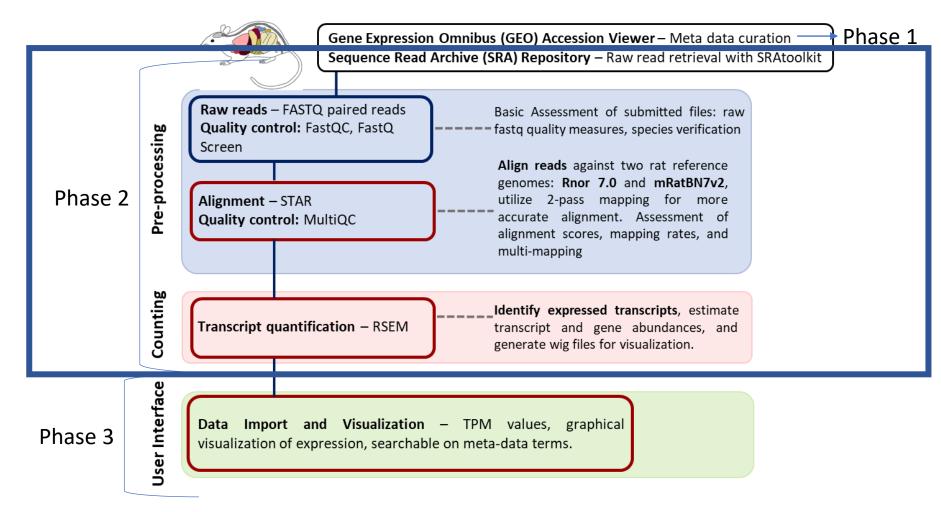
## Summary of Curation Status

- Metadata for 101 studies have been loaded to the database via the new Expression Curation Tool.
- 529 categorized for future curation.
- 24 categorized not for curation (due to lack of information or species).
- We have notified GEO of 15 publications that are associated with GEO Accession IDs but not displayed on the interface.
- We have connected to 21 authors for data clarifications on aspects such as strain, data type, sample to experimental condition relationship.





## Data Re-mapping



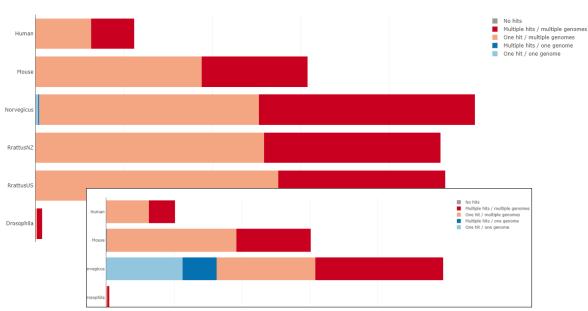
Dobin A, Davis C, Schlesinger F, Drenkow J, Zaleski C, Jha S, Batut P, Chaisson M, Gingeras T: STAR: ultrafast universal RNA-seq aligner. Bioinformatics 2012, Jan:29(1): 15-21

Li B, Dewey C: RSEM: accurate transcript quantification from RNA-Seq data with or without a reference genome. BMC Bioinformatics 2011 (12), 323

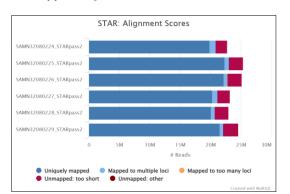




### Data Remapping QC



Wingett SW and Andrews S. FastQ Screen: A tool for multi-genome mapping and quality control [version 2; referees: 4 approved]. F1000Research 2018, 7:1338



RSEM: Mapped reads

SAMN32080224

SAMN32080225

SAMN32080227

SAMN32080228

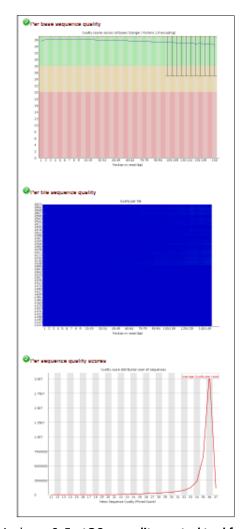
SAMN32080229

0 2.5M 5M 7.5M 10M 12.5M 15M 17.5M 20M # Reads

# Aligned uniquely to a gene Aligned to multiple genes Filtered due to too many alignments Unalignable reads

Created with Multicic

Ewels P, Magnusson M, Lundin S, Käller M: **MultiQC: Summarize analysis results for multiple tools and samples in a single report.** *Bioinformatics* (2016)



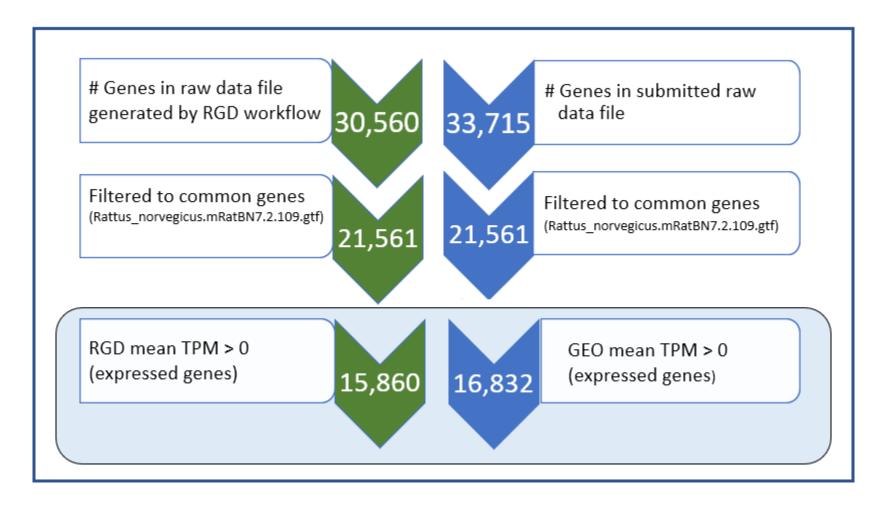
Andrews S. **FastQC: a quality control tool for high throughput sequence data**. 2010. Available from:

https://www.bioinformatics.babraham.ac.uk/projects/fastqc





# Data Re-mapping Evaluation

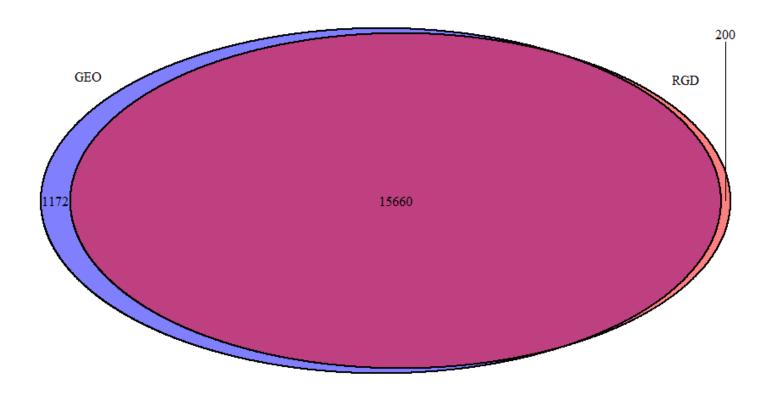


Li X: GSE220261 Available from: <a href="https://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE220261">https://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE220261</a>





# Data Re-mapping Evaluation

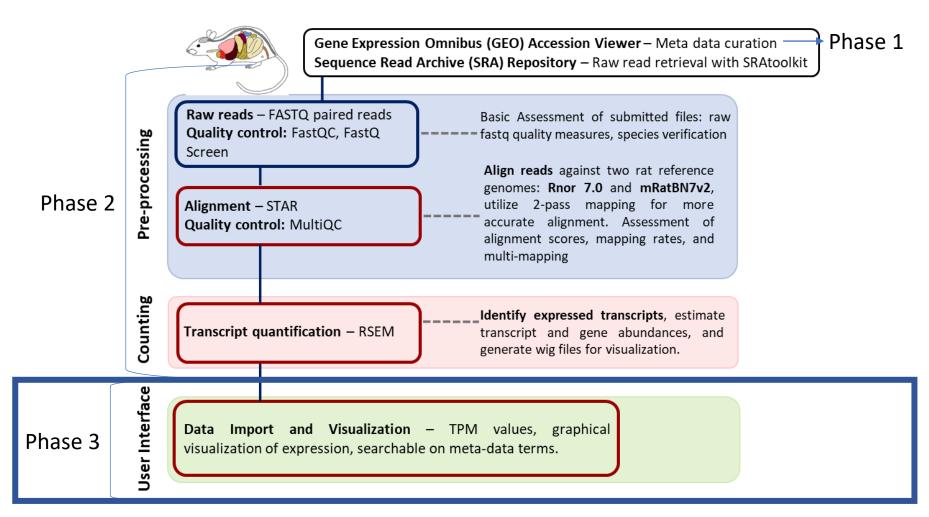


Li X: **GSE220261** Available from: <a href="https://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE220261">https://www.ncbi.nlm.nih.gov/geo/query/acc.cgi?acc=GSE220261</a>





### Data Re-mapping



Dobin A, Davis C, Schlesinger F, Drenkow J, Zaleski C, Jha S, Batut P, Chaisson M, Gingeras T: STAR: ultrafast universal RNA-seq aligner. Bioinformatics 2012, Jan:29(1): 15-21

Li B, Dewey C: RSEM: accurate transcript quantification from RNA-Seq data with or without a reference genome. BMC Bioinformatics 2011 (12), 323





#### JBrowse2



https://www.alliancegenome.org/gene/RGD:628837/#expression





### Acknowledgments

#### **Rat Genome Database**

Principal Investigator
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<u>Project Manager</u> Jennifer Smith, MSc

Research Scientists and Curators

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Mindy Dwinell, PhD

<u>Bioinformatics Manager</u>
Jeff De Pons, BSc

**Developers** 

Adam Gibson, BSc Akhilanand Kundurthi MSc Logan Lamers, BA Jyothi Thota, MSc Marek Tutaj, MSc

<u>Database Administrator</u> Stacy Zacher, MSc

<u>Systems Administrator</u> Kent Brodie, MSc

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And to the researchers who contribute data and faithfully use our website and data!





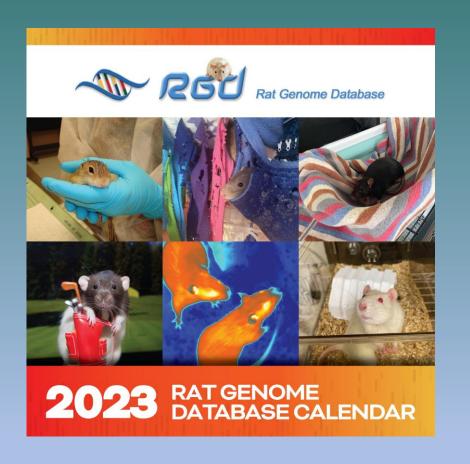
GLOBAL CORE BIODATA RESOURCE







#### Thank you



Special CTC-RG meeting attendee deadline extension is October 13th, 2023

Please send your high-resolution images to RGD.Community@mcw.edu





