

PhenoMiner 2.0: Enhancements in RGD's quantitative phenotype measurement data mining tool improve usability

Jennifer R. Smith

International Mammalian Genomics Conference

July 20, 2022

PhenoMiner 2.0: Enhance RGD's
quantitative phenotype annotation data
minimize the usability

Coming Soon!

Jennifer R. Smith

International Mammalian Genomics Conference

July 20, 2022

A brief introduction to RGD's PhenoMiner

- PhenoMiner is a tool for ontology-based storage and mining of quantitative phenotype data for the laboratory rat
- PhenoMiner includes both data from high-throughput phenotyping projects (standardized) and scientific literature (unstandardized).
- To allow comparisons across studies as well as flexible and intelligent querying, PhenoMiner uses ontologies to express
 - *What was measured* (Clinical Measurement Ontology)
 - *How it was measured* (Measurement Method Ontology)
 - *Under what conditions it was measured* (Experimental Condition Ontology)
 - *In what animals it was measured* (Rat Strain Ontology)
- The Vertebrate Trait Ontology is also used to group related measurements within a study.
- PhenoMiner can be accessed through the Phenotypes and Models tab in the menu at the top of any RGD page or button on RGD's home page.

Limitations of the PhenoMiner 1.0 User Interface

- “The process for selecting records to view is not intuitive.”
- “Once I’ve selected terms it’s hard to change my selections.”
- “I can only view results for one measurement at a time even if I choose terms that use the same units.”
- “I can see the information at the top of the result page but I can’t do anything with it—there’s no way to filter my results on that page.”
- “What? I can sort the results in the table?”
- “Oh, but when I sort the table it doesn’t change anything in the graph. That still appears to be ordered randomly!”
- Coloring the bars in the graph by the experimental conditions is nice but I’d like to choose what they are colored by.”

PhenoMiner 2.0: More intuitive selection and modification

PhenoMiner Database
Rat Strains Selection
• Select 1 or more Rat Strains from the list below.

Select values from categories of interest and select "Generate Report" to build report

Matching Records **2012**

Rat Strains
Search for data related to one or more rat strains.
Examples: congenic strain, ACI, BN
Edit Strains

- SR/Jr (71)
- SR/JrHsd (48)
- SS/JrHsdMcwi (1893)

Additional Options...

- Limit By Clinical Measurements
- Limit By Experimental Conditions
- Limit By Measurement Methods
- I'm Done..
- Generate Report

* If you would like to sh

Select Rat Strains Cancel

• The landing page of the original PhenoMiner was designed to be flexible

Select any ontology

Navigate through the tree?

Go to another page with further options

Back and forth between ontology tree selection pages and "where do you want to go next?" pages

PhenoMiner 2.0: More intuitive selection and modification

PhenoMiner Database Clear

Select values from categories of interest and select **"Generate Report"** to build report

Rat Strains
Search for data related to one or more rat strains.

Clinical Measurements
Query by clinical measurement.

Measurement Methods
Filter results by Measurement method.

Experimental Conditions
Filter based condition.

Generate Report

Rat Strain Selection

Ex: congenic strain, ACI, WKY

select ACI(517)

select ACI-LyStbg-Kyo/Kyo(66)

select ACI.COP-(D10Mgh8-D10Rat4)/Shul(2)

select ACI.COP-(D3Mgh16-D3Rat119)/Shul(2)

select ACI.COP-(D3Rat130-D3Rat114)/Shul(2)

select ACI.COP-(D6Rat80-D6Rat146)/Shul(3)

select ACI.FHH-(D17Rat117-D17Arb5)(D17Rat180-D17Rat51)/Eur(20)

select ACI.FHH-(D1Mit18-D1Mit8)(D14Mit11-D14Hmgc14b)(D14Rat65-D14Rat90)/Eur(8)

select ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(3)

select ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(9)

select ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/EurMcwi(6)

select ACI.FHH-(D1Mit18-D1Rat90)(D3Got102-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)

select ACI.FHH-(D1Mit18-D1Rat90)(D3Rat6-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)


select ACI.FHH-(D1Mit18-D1Rat90)(D3Rat84-D3Rat59)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(6)




StrainsClinical MeasurementsMeasurement MethodsExperimental Conditions

chromosome altered(23471)

- coisogenic strain(640)
- congenic strain(6140)
- consomic strain(19370)
- inbred strain(23376)
- mutant strain(11061)
- outbred strain(4573)
- recombinant inbred strain(2695)
- segregating inbred strain(460)
- transgenic strain(1208)

- In PhenoMiner 2.0 all of the components are on a single page.
- The Strain Ontology loads automatically but you can still start with any of the ontologies by using the tabs in the lower right panel.
- As selections are made in the bottom panels, they appear in the boxes at the top.


Rat Genome Database
<http://rgd.mcw.edu>


DEPARTMENT OF
**BIOMEDICAL
ENGINEERING**
 MEDICAL
COLLEGE
OF WISCONSIN
 MARQUETTE
UNIVERSITY

PhenoMiner 2.0: More intuitive selection and modification

PhenoMiner 2.0
Select value

Rat Strain Selection

Ex: congenic strain, ACI, WKY

- ACI(517)
- ACI-Lystbg-Kyo/Kyo(66)
- ACI.COP-(D10Mgh8-D10Rat4)/Shul(2)
- ACI.COP-(D3Mgh16-D3Rat119)/Shul(2)
- ACI.COP-(D3Rat130-D3Rat114)/Shul(2)
- ACI.COP-(D6Rat80-D6Rat146)/Shul(3)
- ACI.FHH-(D17Rat117-D17Arb5)(D17Rat180-D17Rat51)/Eur(20)
- ACI.FHH-(D1Mit18-D1Mit8)(D14Mit11-D14Hmgc14b)(D14Rat65-D14Rat90)/Eur(8)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(9)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/EurMcwi(6)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Got102-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Rat6-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Rat84-D3Rat59)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(6)

Rat Strain Selection
Ex: congenic strain

- ACI(517)
- ACI-Lystbg-Kyo/Kyo(66)
- ACI.COP-(D10Mgh8-D10Rat4)/Shul(2)
- ACI.COP-(D3Mgh16-D3Rat119)/Shul(2)
- ACI.COP-(D3Rat130-D3Rat114)/Shul(2)
- ACI.COP-(D6Rat80-D6Rat146)/Shul(3)
- ACI.FHH-(D17Rat117-D17Arb5)(D17Rat180-D17Rat51)/Eur(20)
- ACI.FHH-(D1Mit18-D1Mit8)(D14Mit11-D14Hmgc14b)(D14Rat65-D14Rat90)/Eur(8)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(9)
- ACI.FHH-(D1Mit18-D1Rat90)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/EurMcwi(6)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Got102-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Rat6-D3Got149)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Mcwi(3)
- ACI.FHH-(D1Mit18-D1Rat90)(D3Rat84-D3Rat59)(D14Mit11-D14Rat33)(D14Rat65-D14Rat90)/Eur(6)

<http://rgd.mcw.edu>

Prominent Search box

Alphabetical list of terms

PhenoMiner 2.0: More intuitive selection and modification

PhenoMiner Database
Select values from categories of interest and select "Generate Report" to build report

Rat Strain Selection
Search for data related to one or more rat strains.

SS/Jr

select SS/Jr(3110)
select SS/Jr(520)
select SS/Jr.SR/Jr(149)
select SS/Jr.SR/Jr (chr 7)(75)
select SS/Jr.SR/Jr (chr 13)(17)
select SS/Jr.SR/Jr (chr 3)(83)
select SS/Jr.SR/Jr (chr 9)(4)
select SS.SR-(D13N1-D13Mit1)/Jr(2)
select SS.SR-(Sy2-D13Mit1)/Jr(2)

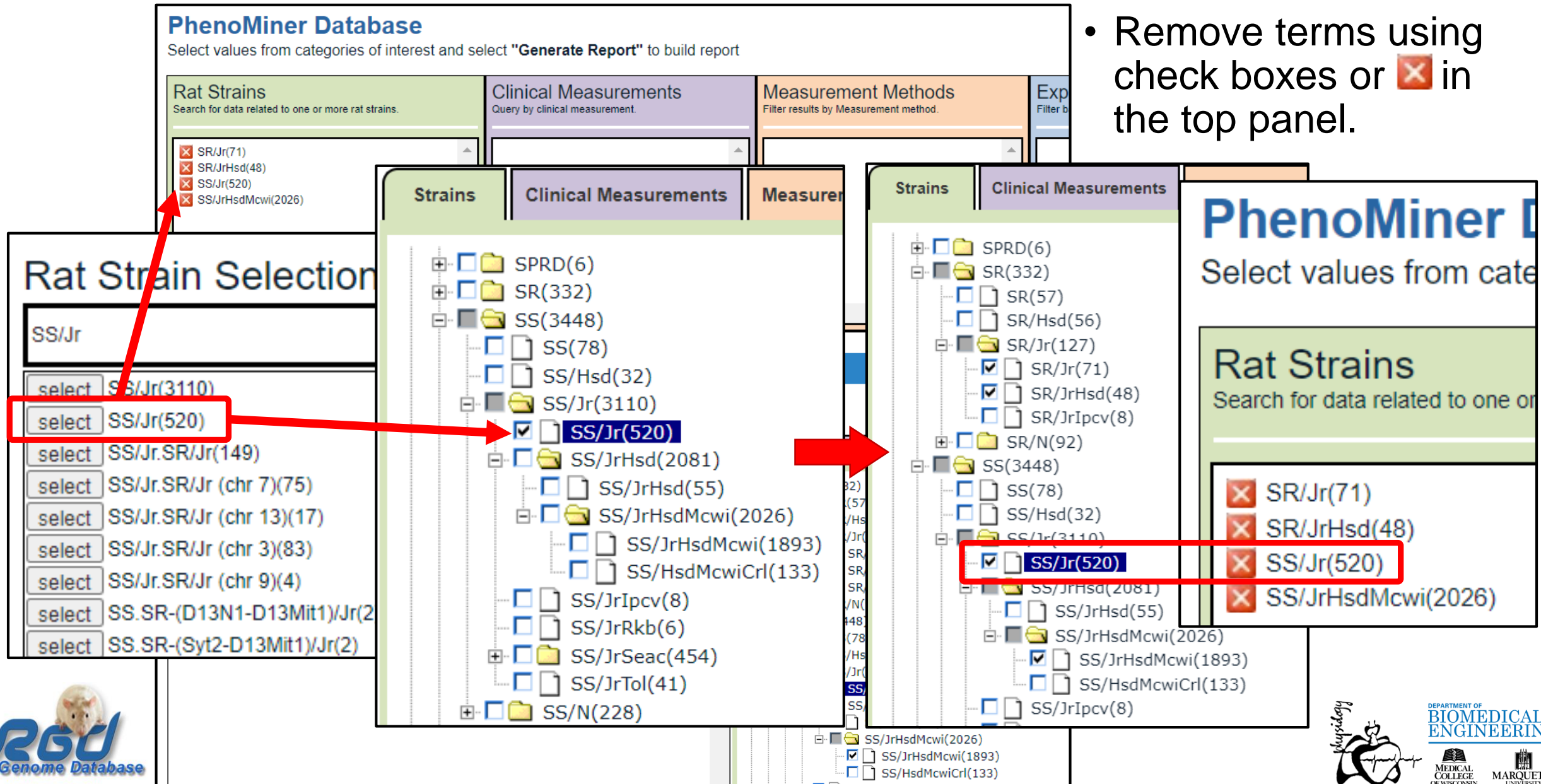
Strains | **Clinical Measurements** | **Measurements**

- SPRD(6)
- SR(332)
- SS(3448)
 - SS(78)
 - SS/Hsd(32)
 - SS/Jr(3110)
 - SS/Jr(520)
 - SS/JrHsd(2081)
 - SS/JrHsd(55)
 - SS/JrHsdMcwi(2026)
 - SS/JrHsdMcwi(1893)
 - SS/HsdMcwiCr1(133)
 - SS/JrIpcv(8)
 - SS/JrRkb(6)
 - SS/JrSeac(454)
 - SS/JrTol(41)
 - SS/N(228)

- Entering a term in the search box narrows the list below it.
- “Select” puts the term into the box at the top and opens that region in the ontology tree in the bottom right panel making it easy to refine the selection, add terms, etc.

PhenoMiner 2.0: More intuitive selection and modification

- Remove terms using check boxes or  in the top panel.



The screenshot illustrates the PhenoMiner 2.0 interface, showing the process of selecting and modifying rat strains. The interface is divided into several panels:

- PhenoMiner Database:** The top panel, containing search filters for Rat Strains, Clinical Measurements, Measurement Methods, and Experimental Conditions. A list of selected strains is shown with red 'X' icons for removal: SR/Jr(71), SR/JrHsd(48), SS/Jr(520), and SS/JrHsdMcwi(2026).
- Rat Strain Selection:** A panel on the left showing a search for "SS/Jr" and a list of selection options. The option "select SS/Jr(520)" is highlighted with a red box. A red arrow points from this box to the "SS/Jr(520)" entry in the main tree view.
- Strains Tree View:** A central tree view showing a hierarchy of rat strains. The "SS/Jr(520)" entry is selected and highlighted with a blue background and a checkmark. A red arrow points from this entry to the "SS/Jr(520)" entry in the right-hand panel.
- PhenoMiner Database (Right Panel):** A panel on the right showing a search for "SS/Jr" and a list of selection options. The option "select SS/Jr(520)" is highlighted with a red box. A red arrow points from this box to the "SS/Jr(520)" entry in the main tree view.

The interface also includes a "Rat Strain Selection" panel on the left, a "Strains" panel in the center, and a "PhenoMiner Database" panel on the right. The "Rat Strain Selection" panel shows a search for "SS/Jr" and a list of selection options. The "Strains" panel shows a tree view of rat strains, with "SS/Jr(520)" selected. The "PhenoMiner Database" panel shows a search for "SS/Jr" and a list of selection options, with "SS/Jr(520)" highlighted.

PhenoMiner 2.0: More intuitive selection and modification

- All selections are made within the

Rat Strains
Search for data related to one or more rat strains.

- SR/Jr(42)
- SR/Jr(32)
- SR/JrHsd(7)
- SS/JrHsdMcwi(252)
- SS/JrHsdMcwi(229)

Clinical Measurements
Query by clinical measurement.

Measurement Methods
Filter results by Measurement method.

Experimental Conditions
Filter based condition.

Rat Strains
Search for data related to one or more rat strains.

- SR/Jr(32)
- SR/Jr(22)
- SR/JrHsd(7)
- SS/JrHsdMcwi(163)
- SS/JrHsdMcwi(141)

Clinical Measurements
Query by clinical measurement.

- mean arterial blood pressure(127)
- mean arterial blood pressure(77)
- diastolic blood pressure(19)
- systolic blood pressure(29)
- systolic blood pressure(28)
- heart rate(90)
- heart rate(46)

Measurement Methods
Filter results by Measurement method.

- tail cuff plethysmography(9)
- radiotelemetry(44)
- radiotelemetry(8)
- intra-aortic abdominal radiotelemetry(18)
- intra-aortic abdominal radiotelemetry(14)
- intra-aortic abdominal radiotelemetry via femoral cannulation(4)
- intra-aortic thoracic radiotelemetry via carotid cannulation(18)
- vascular indwelling catheter method(117)
- vascular indwelling catheter method(41)
- vascular fluid filled catheter(70)

Experimental Conditions
Filter based condition.

- furosemide(25)
- control condition(35)
- vehicle control condition(3)
- controlled sodium content diet(136)
- controlled sodium chloride content diet(14)

Clinical Measurement

Ex: heart rate, blood cell count

- select FAPGG metabolism-surface area p
- select absolute change in blood pH(18)
- select absolute change in body temperatu
- select absolute change in heart rate(56)
- select absolute change in mean arterial b
- select absolute change in partial pressure
- select absolute change in partial pressure
- select absolute change in plasma renin a
- select acetylcholine response/sensitivity r
- select acetylcholine-induced blood vesse
- select in a pre-constricted blood vessel(1
- select adrenal angiotensin II type 1 recep
- select adrenal gland molecular compositi
- select adrenal gland morphological meas
- select adrenal protein/peptide compositi

Generate Report ←

Experimental Condition Selection

Ex: diet, atmosphere composition

- select 17 beta-estradiol(1)
- select NG-nitroarginine methyl ester(3)
- select acetylcholine(4)
- select activity(36)
- select air carbon dioxide content(12)
- select air oxygen content(12)
- select amino acid(3)
- select anesthetic/analgesic(4)
- select angiotensin II(5)
- select angiotensin(5)
- select antibacterial agent(10)
- select anticonvulsant(25)
- select antimicrobial agent(10)
- select bilateral ovariectomy(2)
- select chemical with specified function(43)
- select chemical with specified structure(30)
- select chemical(45)
- select control condition(31)

Strains	Clinical Measurements	Measurement Methods	Experimental Conditions
			<ul style="list-style-type: none"> chemical with specified structure(30) <ul style="list-style-type: none"> amino acid(3) oil(2) peptide/protein(5) steroid(1) sulfonamide(25) <ul style="list-style-type: none"> <input checked="" type="checkbox"/> furosemide(25) control condition(35) <ul style="list-style-type: none"> <input checked="" type="checkbox"/> control condition(31) sham surgical control condition(1) <input checked="" type="checkbox"/> vehicle control condition(3) controlled atmosphere composition(24) controlled visible light condition(18) diet(146) <ul style="list-style-type: none"> drink(10) forced feeding(10) <input checked="" type="checkbox"/> solid diet(136) <input checked="" type="checkbox"/> controlled content diet(136) radiation exposure(9)

The PhenoMiner 2.0 results display consists of three parts

Phenominer Database Results (170 hits)

How to display a graph ?

Strain	Phenotype	Conditions	Study	Experiment Name	Sex
SS/JrHsdMcwi	heart rate	control condition	PhysGen Respiratory data	heart pumping trait	female
SS/JrHsdMcwi	systolic blood pressure	controlled sodium content diet (0.4 %) (for 16 hours) and furosemide (10 mg/kg)	Moreno C, et al., Physiol Genomics 2003 Nov 11;15(3):243-57.	arterial blood pressure trait	female
SS/JrHsdMcwi	mean arterial blood pressure	controlled sodium content diet (4 %) (between 17 and 36 days)	PhysGen Renal A data	arterial blood pressure trait	male
SR/Jr	systolic blood pressure	controlled sodium content diet (8 %) (between 48 and 55 days) then controlled exposure to ambient light	St. Lezin EM, et al., J Clin Invest 1996 Jan 15;97(2):522-7	arterial blood pressure trait	male
SS/JrHsdMcwi	systolic blood pressure	controlled sodium content diet (0.1 %) (for 28 days) then controlled sodium content diet (8	Cowley AW Jr, et al., Physiol Genomics 2000 Apr 27;2(3):107-15.	arterial blood pressure trait	male

Measurements

HEART PUMPING TRAIT (beats/min)

heart rate (46)

ARTERIAL BLOOD PRESSURE TRAIT (mmHg)

mean arterial blood pressure (77)

systolic blood pressure (28)

diastolic blood pressure (19)

Strains

SR

SR/Jr (29)

SR/JrHsd (29)

SS

SS/JrHsdMcwi (141)

Methods

intra-aortic abdominal radiotelemetry (14)

intra-aortic abdominal radiotelemetry via femoral cannulation (4)

intra-aortic thoracic radiotelemetry via carotid cannulation (18)

radiotelemetry (8)

tail cuff

plethysmography (9)

- The PhenoMiner 2.0 results display consists of three parts:
 - A graph
 - A list of filters
 - A table of results
- If measurements that use more than one unit have been selected, the graph is hidden until the user filters the measurement selection

The PhenoMiner 2.0 results display consists of three parts

Measurements

HEART PUMPING TRAIT (beats/min)

- heart rate (46)

ARTERIAL BLOOD PRESSURE TRAIT (mmHg)

- mean arterial blood pressure (69)
- systolic blood pressure (20)
- diastolic blood pressure (11)

Strains

SR

- SR/Jr (8)

SS

- SS/JrHsdMcwi (23)

Phenominer Database Results (31 hits) Download all records

Remove Filters: All systolic blood pressure diastolic blood pressure

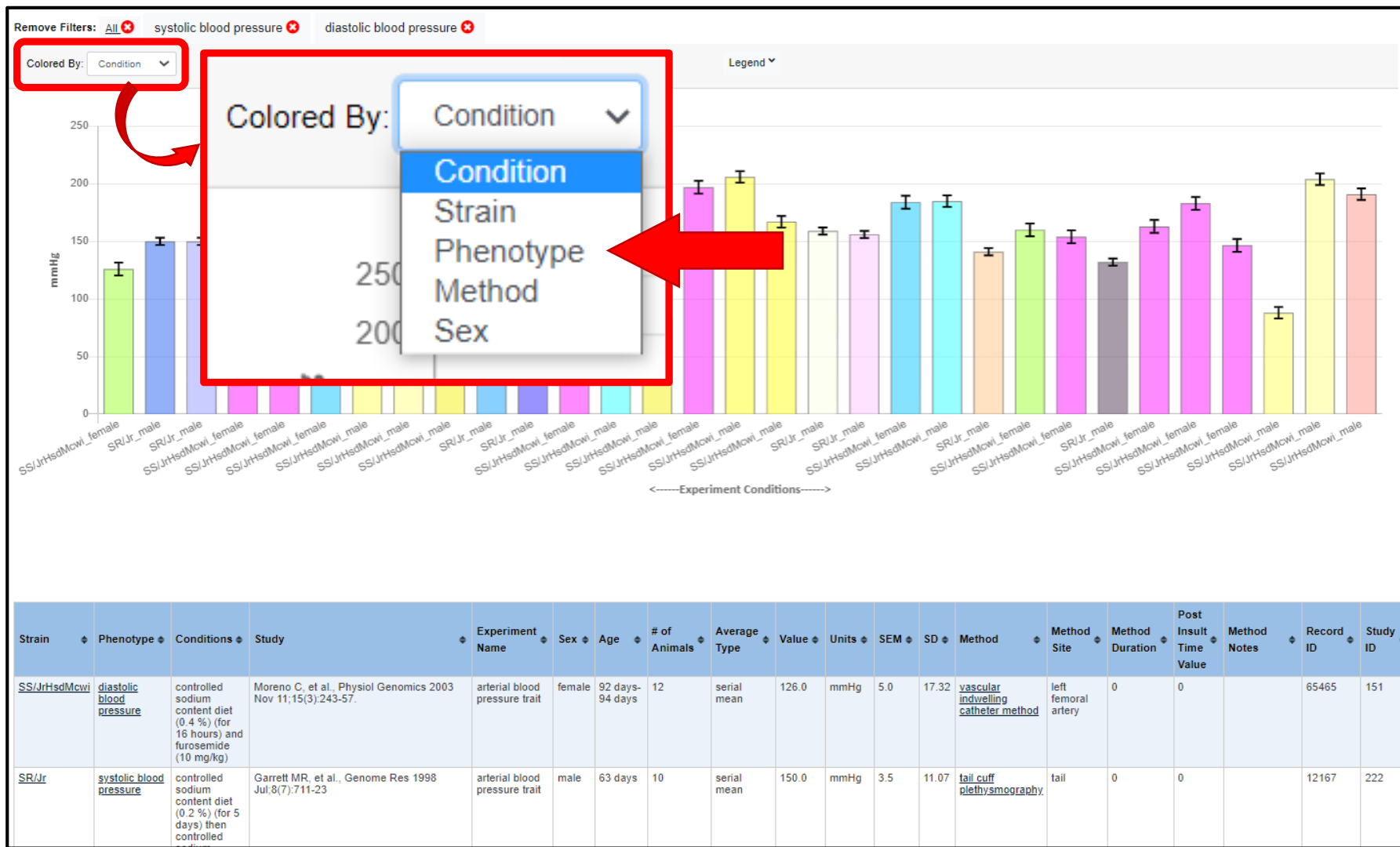
Colored By: Condition Legend

← Experiment Conditions →

Strain	Measurement	Condition	Value (mmHg)
SS/JrHsdMcwi	mean arterial blood pressure	controlled sodium content diet (4%)	~125
SR/Jr	systolic blood pressure	control condition	~125
SR/Jr	diastolic blood pressure	control condition	~145
SS/JrHsdMcwi	mean arterial blood pressure	control condition	~180
SR/Jr	systolic blood pressure	control condition	~140
SR/Jr	diastolic blood pressure	control condition	~140
SS/JrHsdMcwi	mean arterial blood pressure	control condition	~120
SR/Jr	systolic blood pressure	control condition	~150
SR/Jr	diastolic blood pressure	control condition	~115
SS/JrHsdMcwi	mean arterial blood pressure	control condition	~205
SR/Jr	systolic blood pressure	control condition	~140
SR/Jr	diastolic blood pressure	control condition	~140
SS/JrHsdMcwi	mean arterial blood pressure	control condition	~85
SR/Jr	systolic blood pressure	control condition	~195
SR/Jr	diastolic blood pressure	control condition	~205
SS/JrHsdMcwi	mean arterial blood pressure	control condition	~165
SR/Jr	systolic blood pressure	control condition	~155
SR/Jr	diastolic blood pressure	control condition	~155

that filter at the top of the page

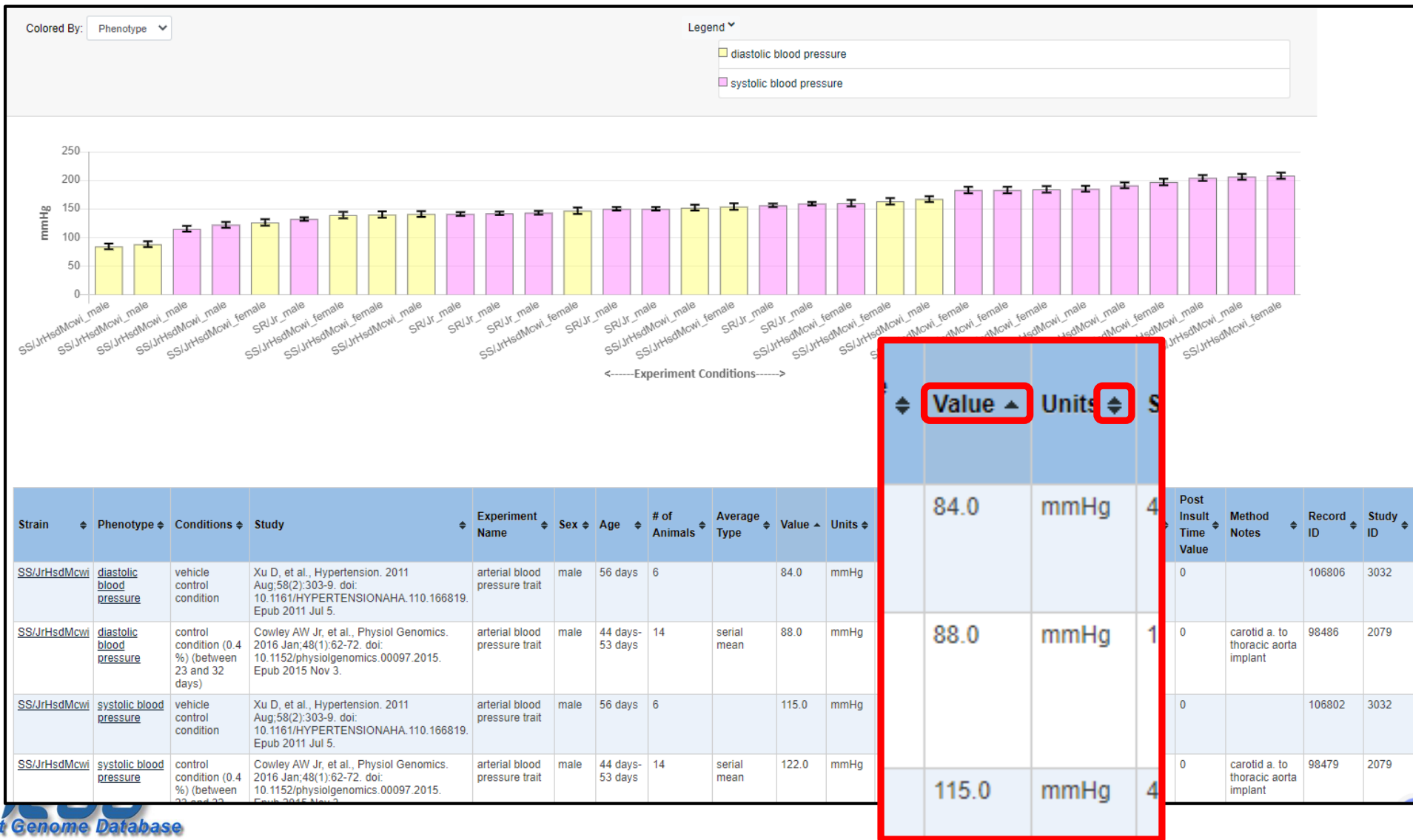
The PhenoMiner 2.0 graph is more interactive: “Colored by” function



The PhenoMiner 2.0 graph is more interactive: “Colored by” function



The PhenoMiner 2.0 graph is more interactive: Sorting the table reorders the bars in the graph



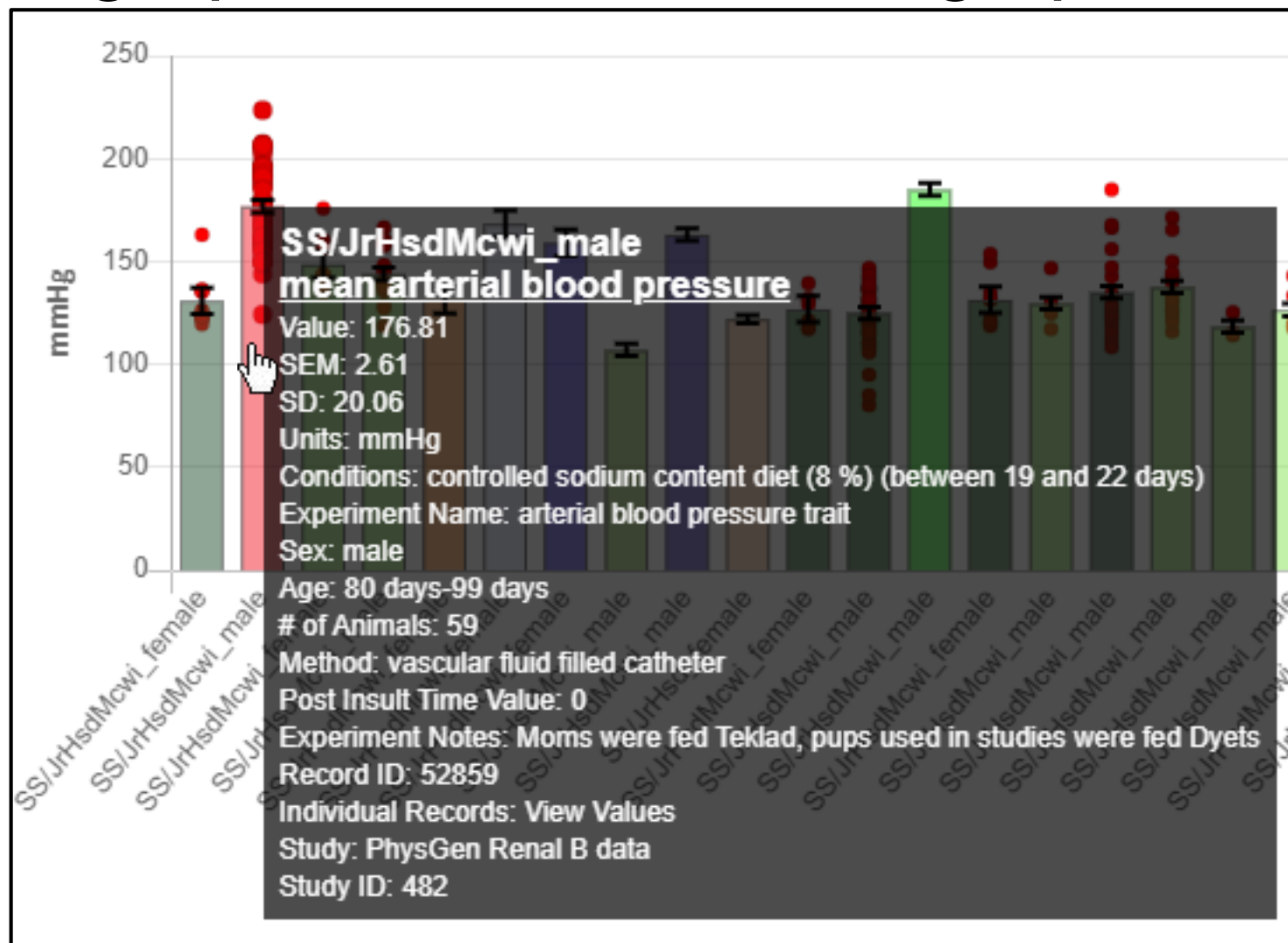
- Using colored by phenotype and sort by value it is easy to see at a glance the mix of diastolic and systolic blood pressure values.

Where data for individual rats has been submitted, the graph shows the average plus the individual values



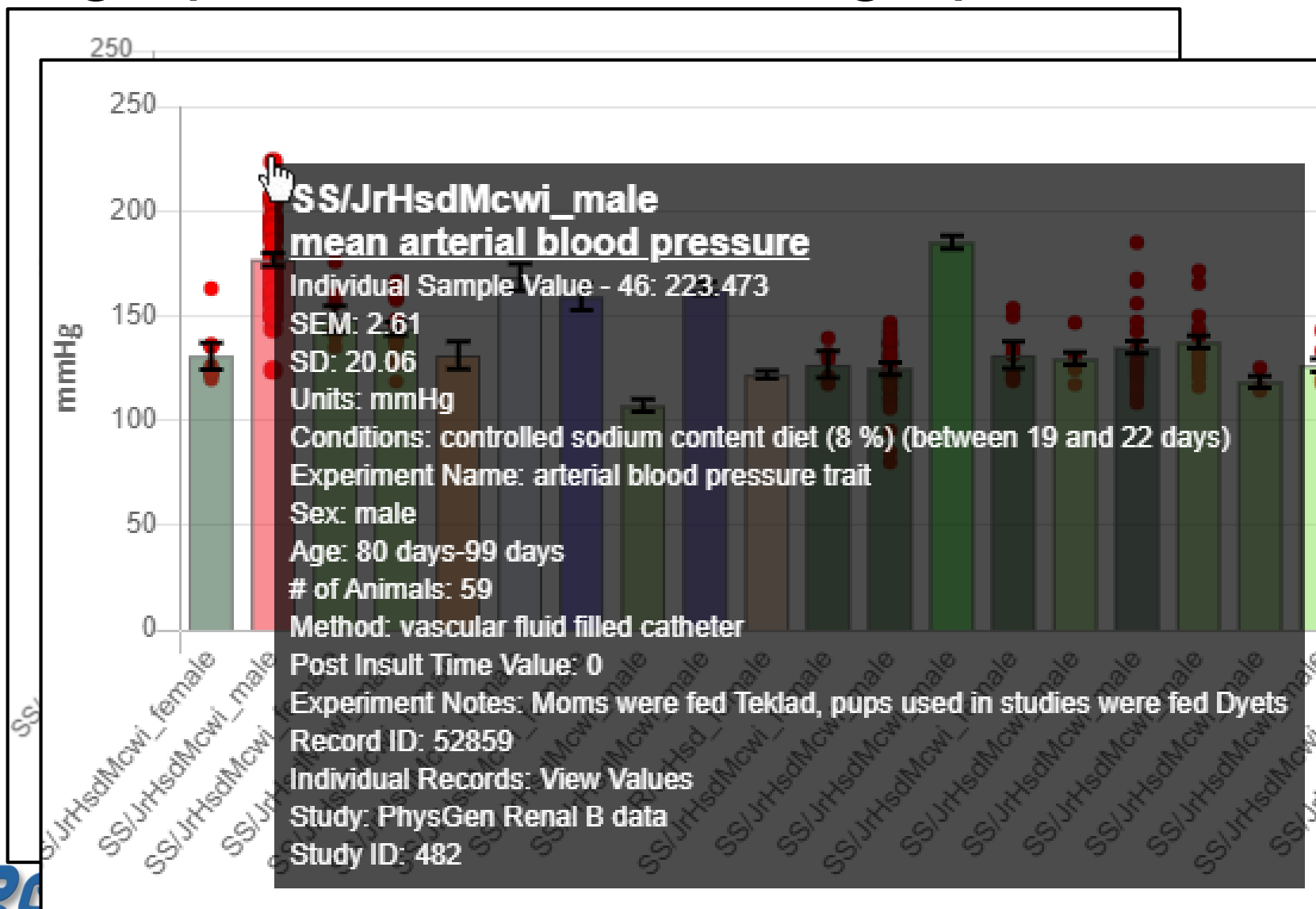
Looking at the graph for mean arterial blood pressure, you can see that some of the bars include red dots indicating the values for individual animals.

Where data for individual rats has been submitted, the graph shows the average plus the individual values



- Mouse over the bar to see a popup with details about the average value for that sample of rats under those conditions.

Where data for individual rats has been submitted, the graph shows the average plus the individual values



- Mouse over the bar to see a popup with details about the average value for that sample of rats under those conditions.
- Mouse over a dot to see the details for that individual rat measurement.

Where data for individual rats has been submitted, the table shows the average with the option to view individual values

The screenshot displays the Rat Genome Database interface. At the top, a bar chart shows mean arterial blood pressure (mmHg) for various rat strains. Below the chart, a table lists experimental details. A red box highlights the 'View Values' button for study ID 481, which opens a pop-up window showing individual sample values. Red arrows indicate the flow of information from the 'View Values' button to the individual values list and then to the 'Individual Value' column in the table below.

Strain	Phenotype	Conditions	Study	Experiment Name	Sex	Age	# of Animals	Formulation
SS/JrHsdMcv	mean arterial blood pressure	controlled sodium content diet (4%) (between 17 and 36 days) then furosemide (10 mg/kg) then controlled sodium content diet (0.4%) (for 2 days)	PhysGen Renal A data	arterial blood pressure trait	female	87 days-120 days	10	
SS/JrHsdMcv	mean arterial blood pressure	controlled sodium content diet (8%) (between 19 and 22	PhysGen Renal B data	arterial blood pressure trait	male	80 days-99 days	59	

Individual Sample Values for Study ID 481:

- F101SS: 119.57
- F107SS: 122.22
- F166SS: 122.76
- F259SS: 122.97
- F102SS: 124.96
- F106SS: 126.31
- F104SS: 135.19
- F108SS: 136.01
- F309SS: 136.34
- F165SS: 163

Data from the query can be downloaded

The screenshot displays the Phenominer Database interface. On the left, there are filter panels for 'Measurements' (Heart Pumping Trait, Arterial Blood Pressure Trait) and 'Strains' (SR, SS). The main area shows 'Phenominer Database Results (170 hits)' with a table of columns: Strain, Phenotype, Conditions, Study, Experiment Name, Sex, Age, # of Animals. A 'Download all records' button is visible in the top right. Below the table, a bar chart shows 'Phenominer Database Results (100 hits)' with filters for 'mean arterial blood pressure', 'systolic blood pressure', and 'diastolic blood pressure'. A 'Download all records' button and a 'Download table view records' button are present in the top right of the chart area. A red box highlights the 'Download all records' button in both the table and chart views.

A button for “Download all records” is displayed on the pages for both unfiltered and filtered queries.

Filtered queries also have an option to download just the table view records.

The RGD Team:

Principal Investigator:

Anne Kwitek, PhD

Co-Investigator:

Mindy Dwinell, PhD

Curation Team:

Jennifer Smith, MSc

Stan Laulederkind, PhD

Tom Hayman, PhD

Shur-Jen Wang, PhD

Mary Kaldunski, MSc

Mahima VEDI, PhD

Wendy Demos, MSc

Morgan Hill, MMP

Monika Tutaj, PhD

Development Team:

Jeff De Pons, BSc

Marek Tutaj, MSc

Jyothi Thota, MSc

Logan Lamers, BSc

Harika S Nalabolu, MSc

Adam Gibson, BSc

Ketaki Thorat, BSc

Thank you!



We gratefully acknowledge our funders:

RGD is funded by grant HL64541 from the National Heart, Lung, and Blood Institute

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

jrsmith@mcw.edu

<https://rgd.mcw.edu>

PhenoMiner 2.0: More intuitive selection and modification

The screenshot shows a hierarchical tree of folders and files. The folders are highlighted in yellow, and the files are shown as white icons. The tree structure is as follows:

- SS(3448)
 - SS(78)
 - SS/Hsd(32)
 - SS/Jr(3110)
 - SS/Jr(520)
 - SS/JrHsd(2081)
 - SS/JrHsd(55)
 - SS/JrHsdMcwi(2026)
 - SS/JrHsdMcwi(1893)
 - SS/HsdMcwiCrl(133)
 - SS/JrIpcv(8)

Two red arrows point from text boxes to the folders SS/Jr(3110) and SS/Jr(520).

3110 = # records for SS/Jr and any substrain below it

520 = # records assigned directly to the strain SS/Jr