

VisRseq: R-based visual analytics software for sequencing data

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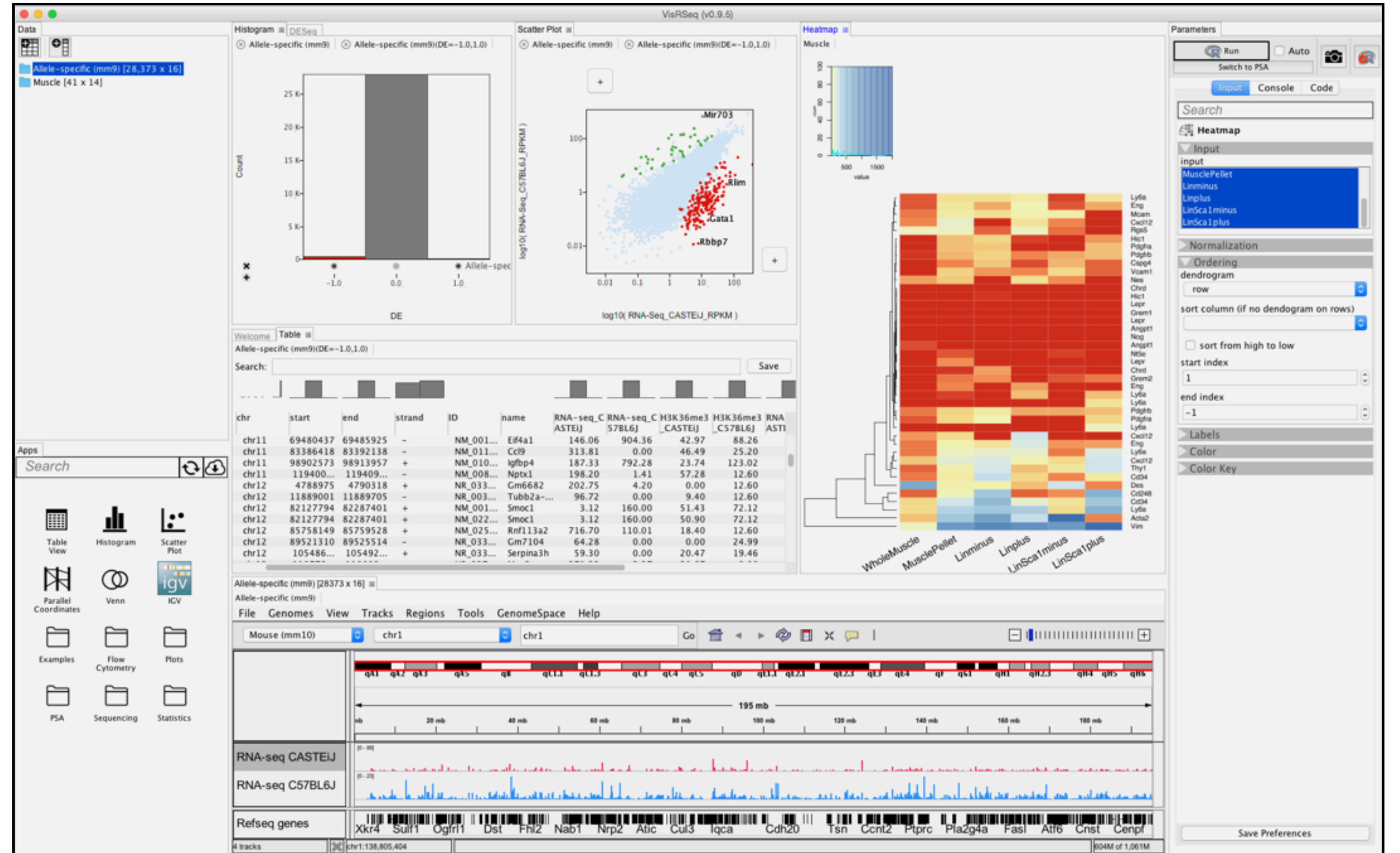
Steven J.M. Jones

Genome Sciences Centre, BC Cancer Agency, Canada



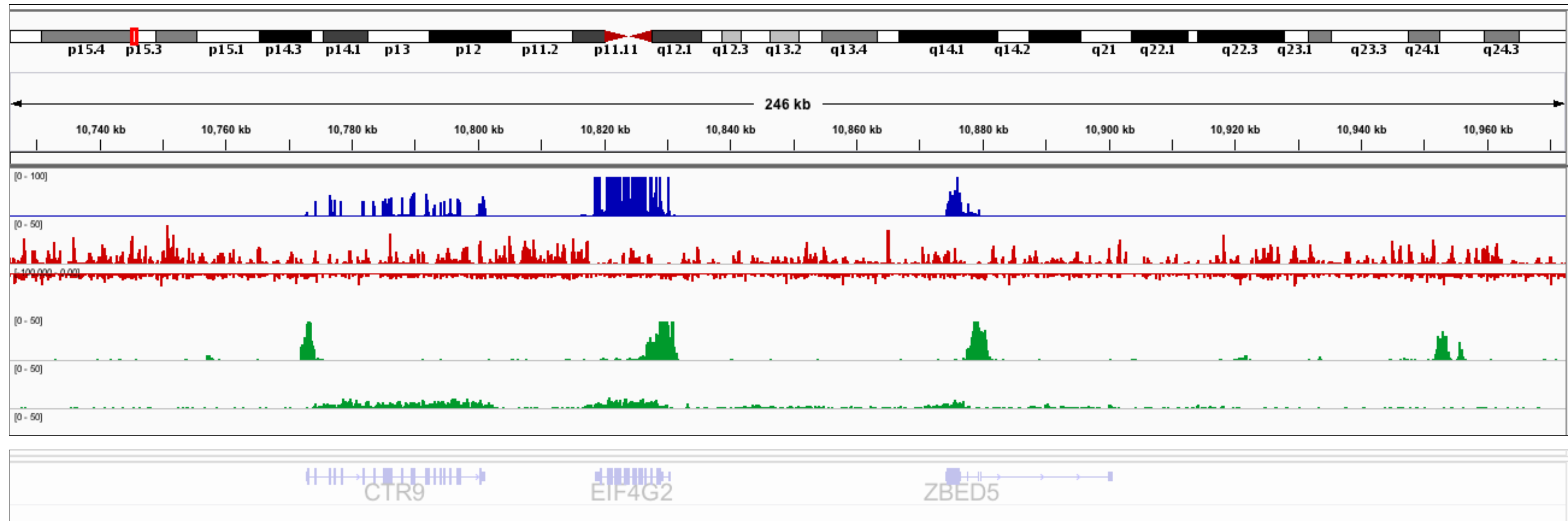
Overview

- introduction
- motivation
- VisRseq
- case study
- future works



introduction

data: sequencing data



- numerical measures for biological properties per genomic location
- RNA-seq, ChIP-seq, Bis-seq, MeDIP, etc.

data: data table

↑ regions ↓

↓ index	chr	start	end	strand	name	ID	Exon Len...
22293	chr11	10562782	10621479	+	MRVI1-AS1	NR_046374	900
22294	chr11	10562782	10621479	+	MRVI1-AS1	NR_034094	623
22295	chr11	10562782	10621479	+	MRVI1-AS1	NR_034093	661
22296	chr11	10579412	10590365	-	LYVE1	NM_0066...	2500
22297	chr11	10594637	10715535	-	MRVI1	NM_0012...	6155
22298	chr11	10594637	10715535	-	MRVI1	NM_0011...	6313
22299	chr11	10594637	10715535	-	MRVI1	NM_0012...	5748
22300	chr11	10594637	10715121	-	MRVI1	NM_0011...	6030
22301	chr11	10594637	10673848	-	MRVI1	NM_0010...	6035
22302	chr11	10594637	10715535	-	MRVI1	NM_1303...	6366
22303	chr11	10772810	10801290	+	CTR9	NM_0146...	4309
22304	chr11	10818592	10830582	-	EIF4G2	NM_0014...	3911
22305	chr11	10818592	10830582	-	EIF4G2	NM_0010...	3797
22306	chr11	10818592	10829543	-	EIF4G2	NM_0011...	4028
22307	chr11	10823013	10823155	-	SNORD97	NR_004403	142
22308	chr11	10874250	10879620	-	ZBED5	NM_0212...	2741
22309	chr11	10874250	10879620	-	ZBED5	NM_0011...	2709
22310	chr11	10879763	10900823	+	ZBED5-AS1	NR_034137	1113
22311	chr11	11292420	11643561	-	GALNT18	NM_1985...	2515
22312	chr11	11373488	11374904	-	CSNK2A3	NM_0012...	1416
22313	chr11	11678197	11678269	-	MIR4299	NR_036184	72
22314	chr11	11862969	11980872	+	USP47	NM_0179...	7777
22315	chr11	11984542	12030186	-	DKK3	NM_0010...	2578



data: data table

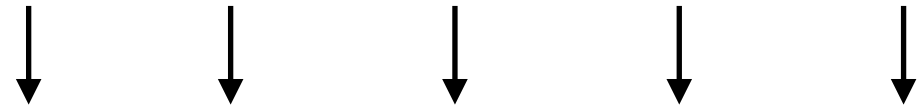
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22293	chr11	10562782	10621479	+	MRVI1-AS1	NR_046374	900
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22297	chr11	10594637	10715535	-	MRVI1	NM_0012...	6155
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22304	chr11	10818592	10830582	-	EIF4G2	NM_0014...	3911
22305	chr11	10818592	10830582	-	EIF4G2	NM_0010...	3797
22306	chr11	10818592	10829543	-	EIF4G2	NM_0011...	4028
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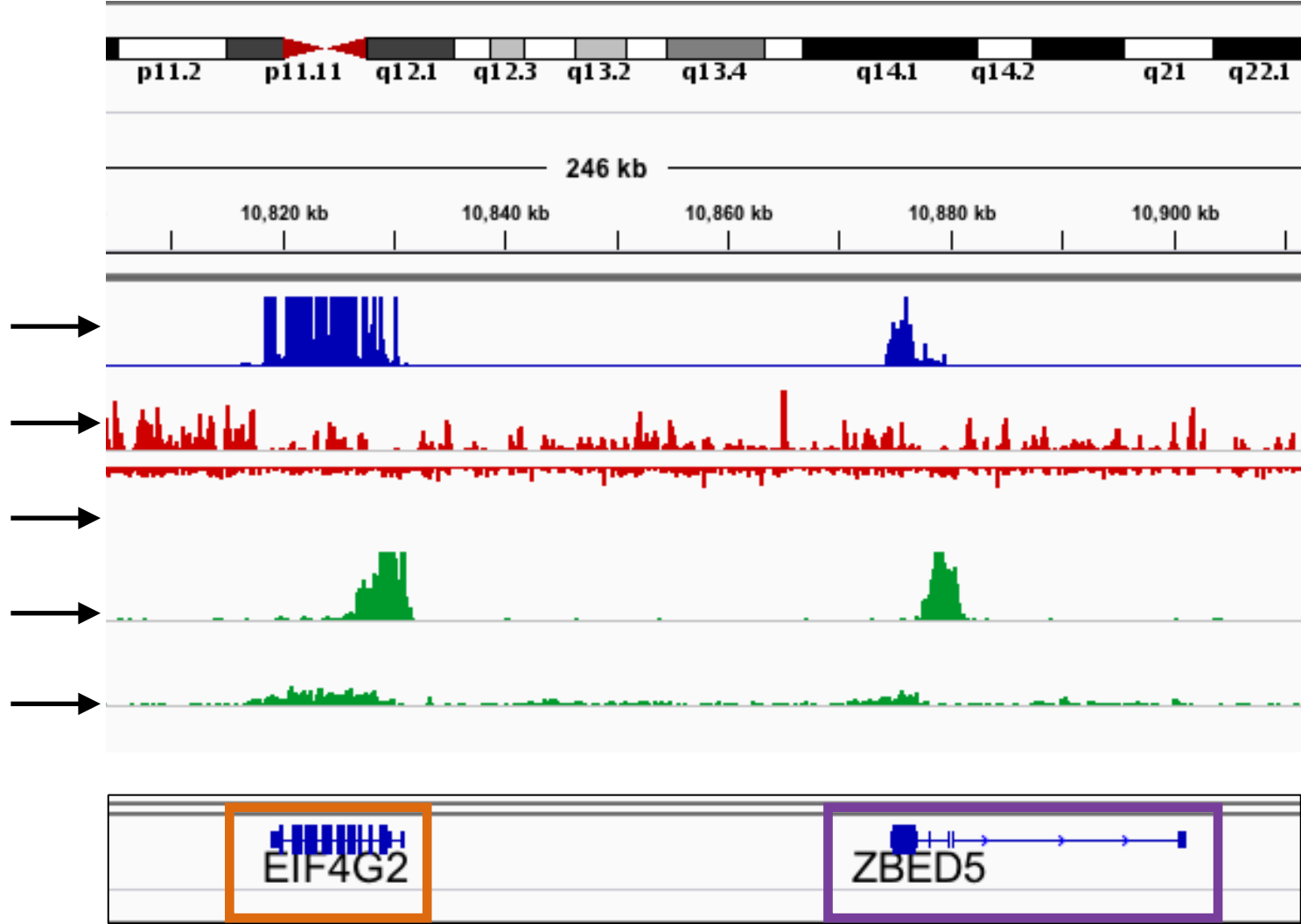
data: data table

sequencing data



regions

↓ index	chr	start	end	strand	name	ID	Exon Len...	RNA-seq	Bis-Seq	MeDIP-seq	H3K4me3	H3K36me3
22293	chr11	10562782	10621479	+	MRVI1-AS1	NR_046374	900	0.138310...	0.375729...	0.326040...	13.93396...	1.071643...
22294	chr11	10562782	10621479	+	MRVI1-AS1	NR_034094	623	0.195220...	0.375729...	0.326040...	13.93396...	1.071643...
22295	chr11	10562782	10621479	+	MRVI1-AS1	NR_034093	661	0.183997...	0.375729...	0.326040...	13.93396...	1.071643...
22296	chr11	10579412	10590365	-	LYVE1	NM_0066...	2500	0.224873...	0.381754...	0.318386...	0.159771...	0.523839...
22297	chr11	10594637	10715535	-	MRVI1	NM_0012...	6155	1.116872...	0.421562...	0.384812...	11.98244...	1.438158...
22298	chr11	10594637	10715535	-	MRVI1	NM_0011...	6313	1.096068...	0.421562...	0.384812...	11.98244...	1.438158...
22299	chr11	10594637	10715535	-	MRVI1	NM_0012...	5748	1.133492...	0.421562...	0.384812...	11.98244...	1.438158...
22300	chr11	10594637	10715121	-	MRVI1	NM_0011...	6030	1.146873...	0.420463...	0.386134...	16.15650...	1.441797...
22301	chr11	10594637	10673848	-	MRVI1	NM_0010...	6035	1.148725...	0.422225...	0.413478...	0.055586...	1.708579...
22302	chr11	10594637	10715535	-	MRVI1	NM_1303...	6366	1.088707...	0.421562...	0.384812...	11.98244...	1.438158...
22303	chr11	10772810	10801290	+	CTR9	NM_0146...	4309	16.63586...	0.377252...	0.300058...	20.88678...	1.924949...
22304	chr11	10818592	10830582	-	EIF4G2	NM_0014...	3911	392.4496...	0.375217...	0.207826...	28.63070...	2.168475...
22305	chr11	10818592	10830582	-	EIF4G2	NM_0010...	3797	391.7547...	0.375217...	0.207826...	28.63070...	2.168475...
22306	chr11	10818592	10829543	-	EIF4G2	NM_0011...	4028	379.5769...	0.348875...	0.222936...	38.06138...	2.314094...
22307	chr11	10823013	10823155	-	SNORD97	NR_004403	142	13.90393...	0.254193...	1.285436...	0.776841...	1.563233...
22308	chr11	10874250	10879620	-	ZBED5	NM_0212...	2741	20.11509...	0.358882...	0.240515...	28.81745...	1.330854...
22309	chr11	10874250	10879620	-	ZBED5	NM_0011...	2709	20.18505...	0.358882...	0.240515...	28.81745...	1.330854...
22310	chr11	10879763	10900823	+	ZBED5-AS1	NR_034137	1113	0.359236...	0.298254...	0.238044...	27.35785...	0.505412...
22311	chr11	11292420	11643561	-	GALNT18	NM_1985...	2515	3.651898...	0.424367...	0.469225...	16.51064...	0.403031...
22312	chr11	11373488	11374904	-	CSNK2A3	NM_0012...	1416	31.65218...	0.417866...	1.022515...	0.111172...	0.423487...
22313	chr11	11678197	11678269	-	MIR4299	NR_036184	72	0.0	0.709756...	0.0	0.194049...	0.0
22314	chr11	11862969	11980872	+	USP47	NM_0179...	7777	19.38955...	0.281544...	0.173758...	14.87724...	1.030983...
22315	chr11	11984542	12030186	-	DKK3	NM_0010...	2578	78.74334...	0.431295...	0.276000...	30.28513...	1.790080...

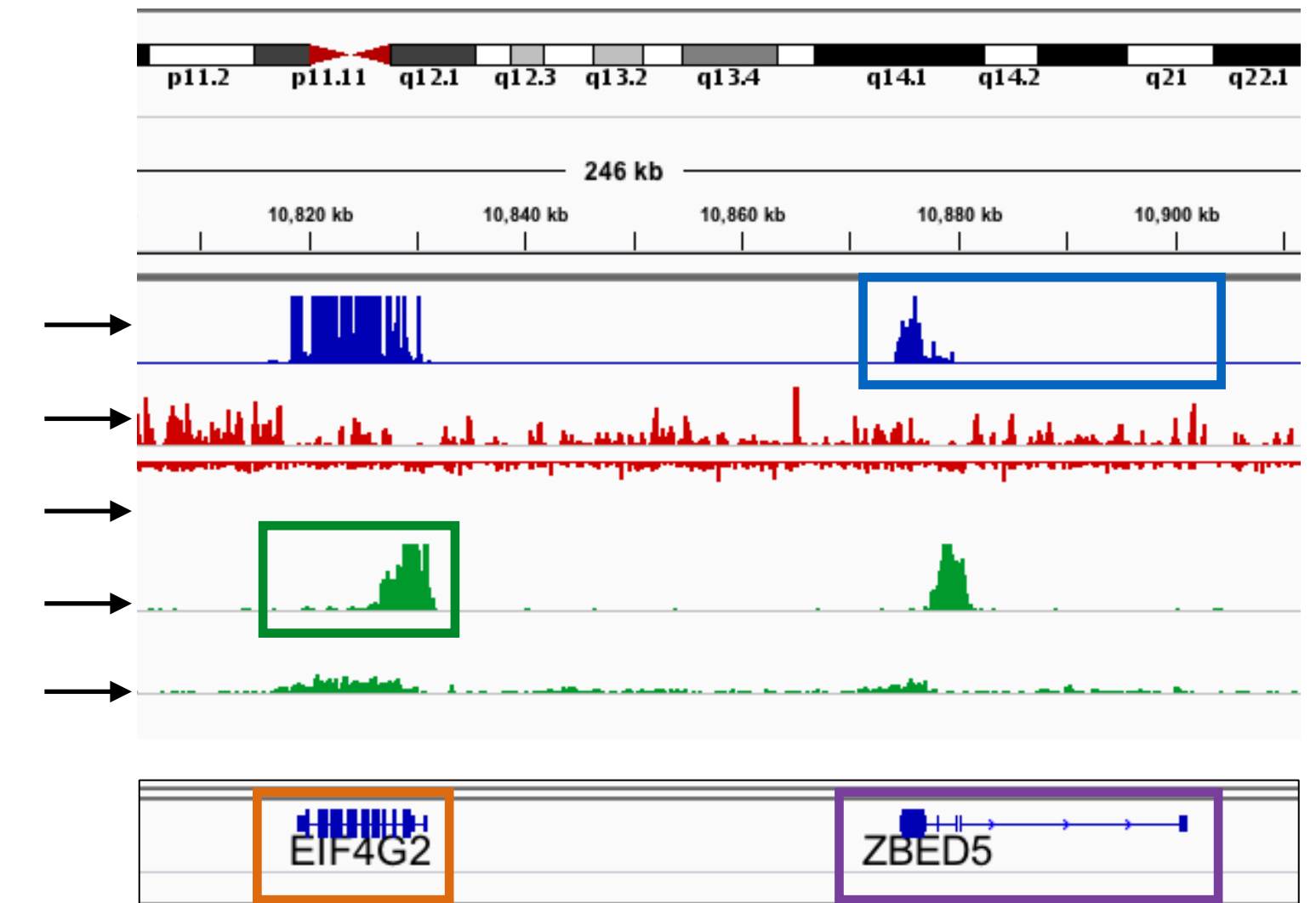
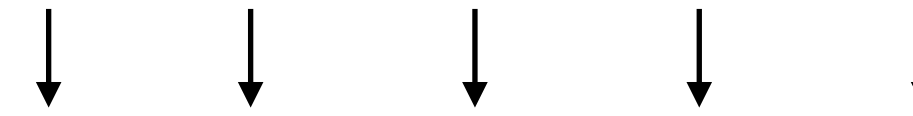


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22313	chr11	11678197	11678269	-	MIR4299	NR_036184	72	0.0	0.709756...	0.0	0.194049...	0.0
22314	chr11	11862969	11980872	+	USP47	NM_0179...	7777	19.38955...	0.281544...	0.173758...	14.87724...	1.030983...
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sequencing data



motivation

motivation

analysis software

motivation

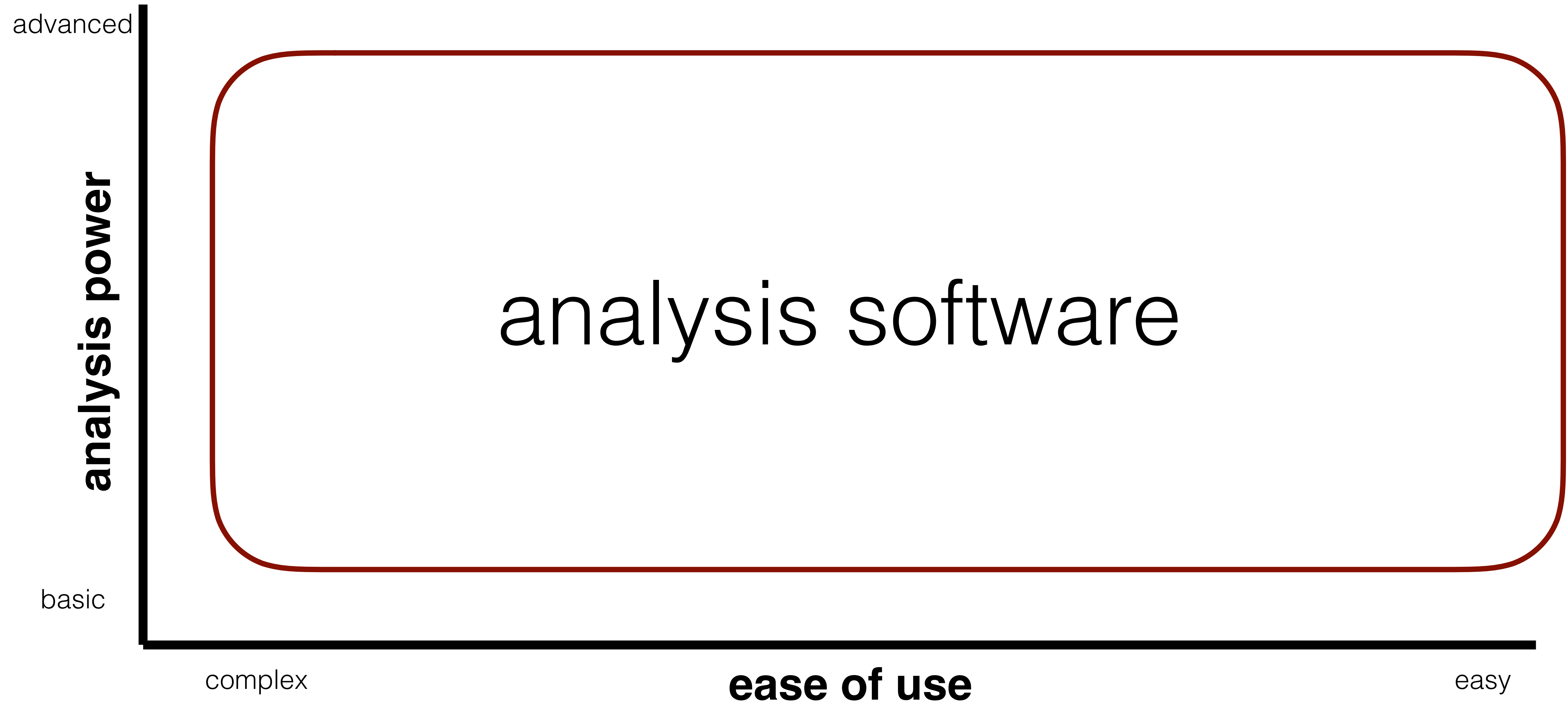
analysis software

complex

ease of use

easy

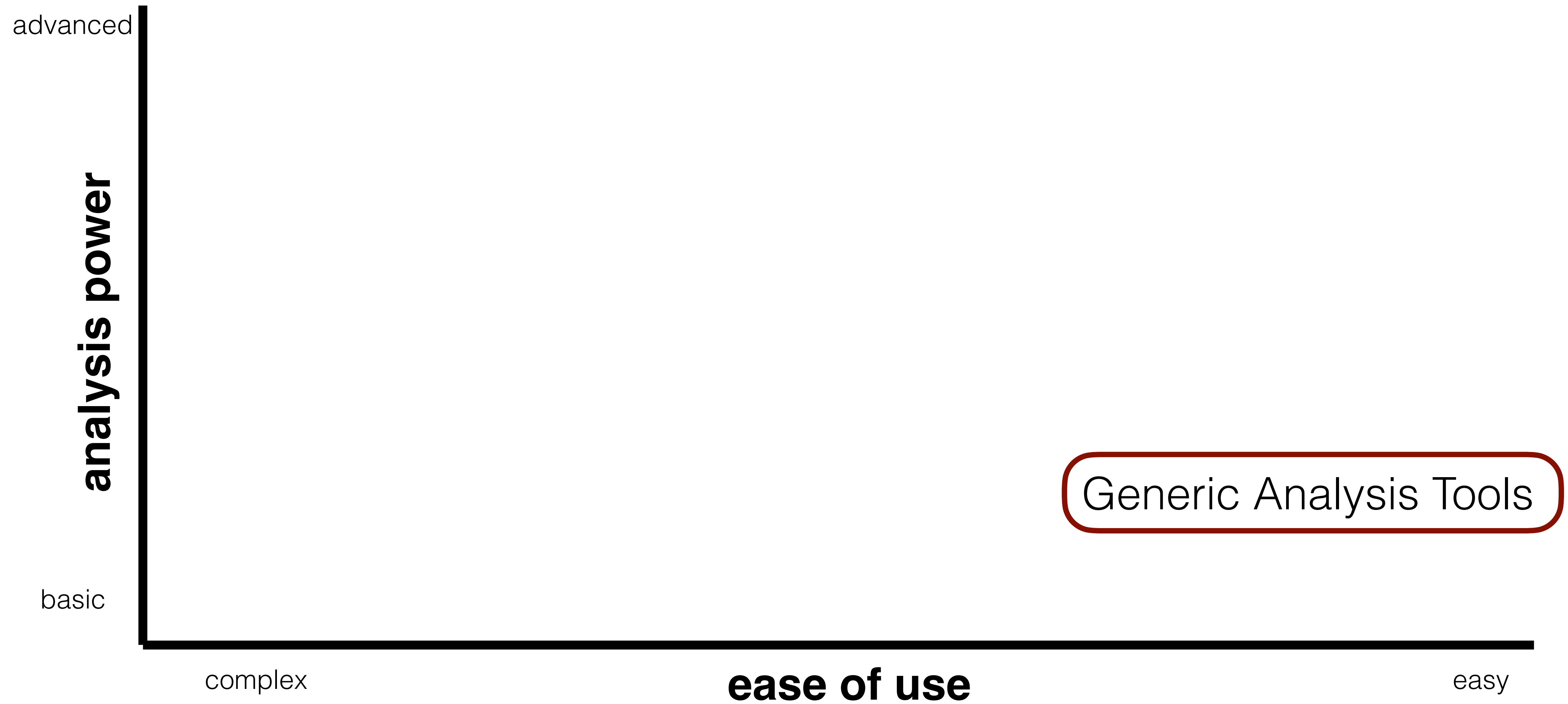
motivation



motivation



motivation



motivation

advanced

analysis power

basic

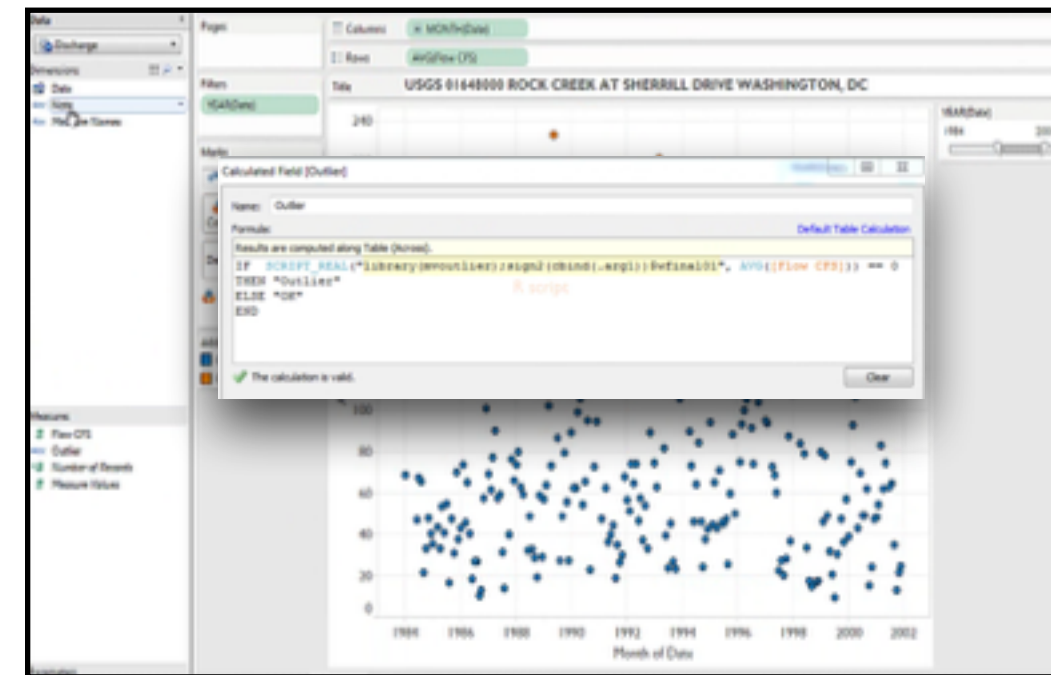
complex

ease of use

easy

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GOTERM_BP GO:0034645	cellular macromolecule biosynthetic process	2.58363459	1.37E-09	42
GOTERM_BP GO:0009059	macromolecule biosynthetic process	2.56246481	1.77E-09	42
GOTERM_BP GO:0010467	gene expression	2.28422179	2.51E-07	39
GOTERM_BP GO:0044267	cellular protein metabolic process	2.09523711	3.58E-06	38
GOTERM_BP GO:0019538	protein metabolic process	1.95086485	4.53E-06	42
GOTERM_BP GO:0044249	cellular biosynthetic process	1.78235807	8.96E-06	47
GOTERM_BP GO:0009058	biosynthetic process	1.70532395	3.13E-05	47
GOTERM_BP GO:0030199	collagen fibril organization	29.7840112	3.04E-04	4
GOTERM_BP GO:0030198	extracellular matrix organization	13.8111342	4.50E-04	5
GOTERM_BP GO:0022900	electron transport chain	5.39395479	6.68E-04	8
GOTERM_BP GO:0043062	extracellular structure organization	8.15514593	0.00319551	5
GOTERM_BP GO:0022904	respiratory electron transport chain	12.0181098	0.00435644	4
GOTERM_BP GO:0043170	macromolecule metabolic process	1.33494519	0.00590397	52
GOTERM_BP GO:0044237	cellular metabolic process	1.22562383	0.00676271	67
GOTERM_BP GO:0044260	cellular macromolecule metabolic process	1.36054073	0.00680266	48
GOTERM_BP GO:0045333	cellular respiration	4.82417083	0.00783823	6
GOTERM_BP GO:0051216	cartilage development	19.7605459	0.00980216	3
GOTERM_BP GO:0008292	acetylcholine biosynthetic process	171.258065	0.0115197	2
GOTERM_BP GO:0009100	glycoprotein metabolic process	5.63348896	0.01164616	5
GOTERM_BP GO:0009987	cellular process	1.1327528	0.01372666	80
GOTERM_BP GO:0006091	generation of precursor metabolites and products	2.59875667	0.01379198	10
GOTERM_BP GO:0032501	multicellular organismal process	1.70689101	0.01607076	21
GOTERM_BP GO:0042136	neurotransmitter biosynthetic process	85.6290323	0.02290813	2
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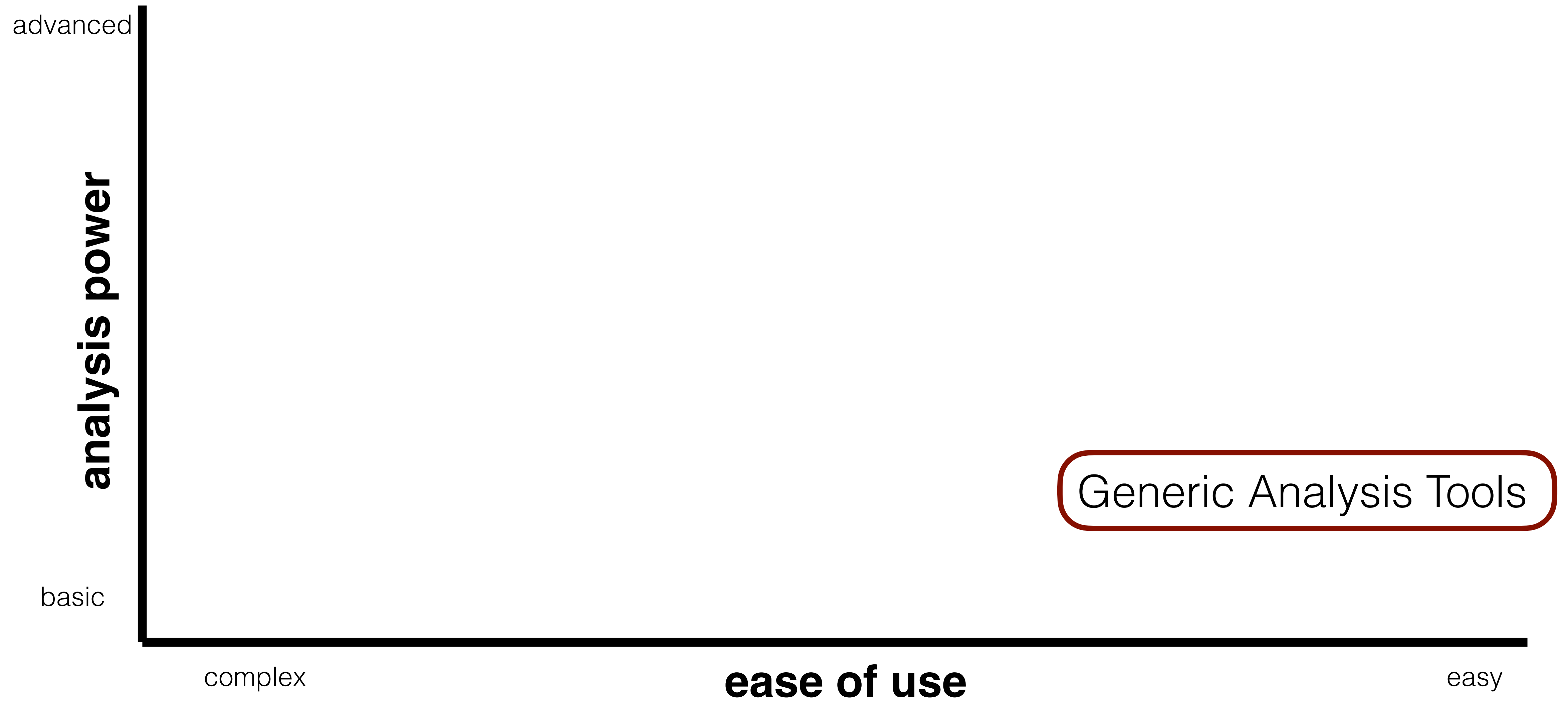
Excel



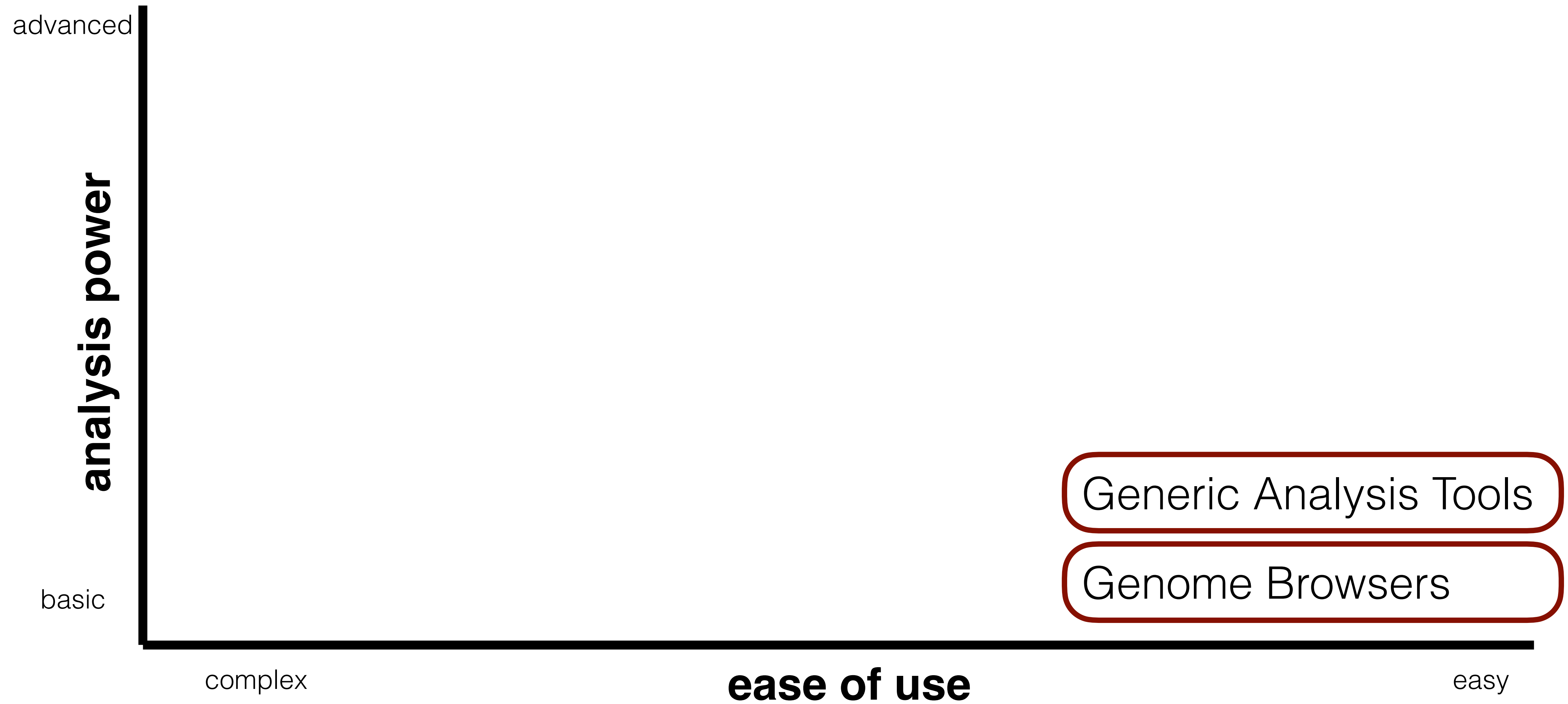
Tableau

Generic Analysis Tools

motivation



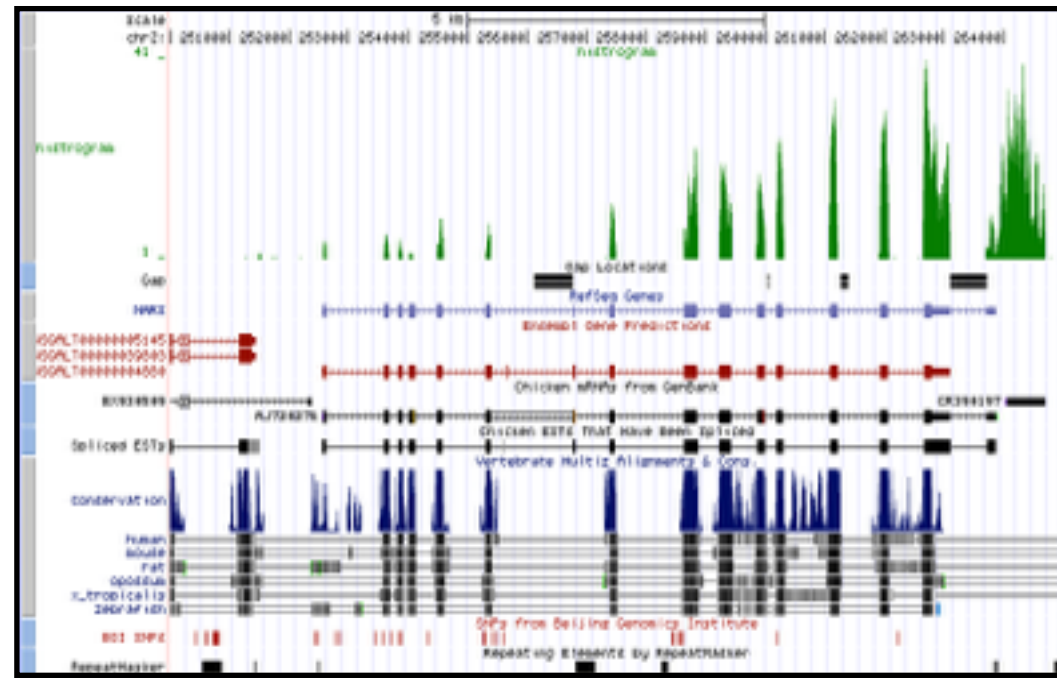
motivation



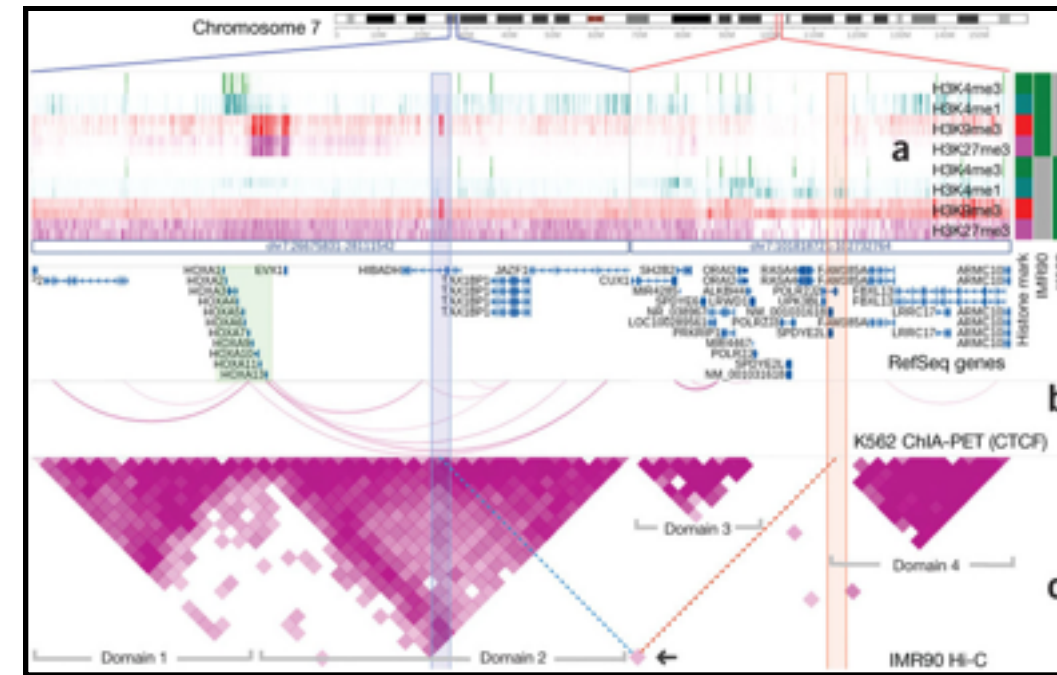
motivation

advanced

analysis power



UCSC Genome Browser



WashU Epigenome Browser



Integrated Genome Viewer

basic

complex

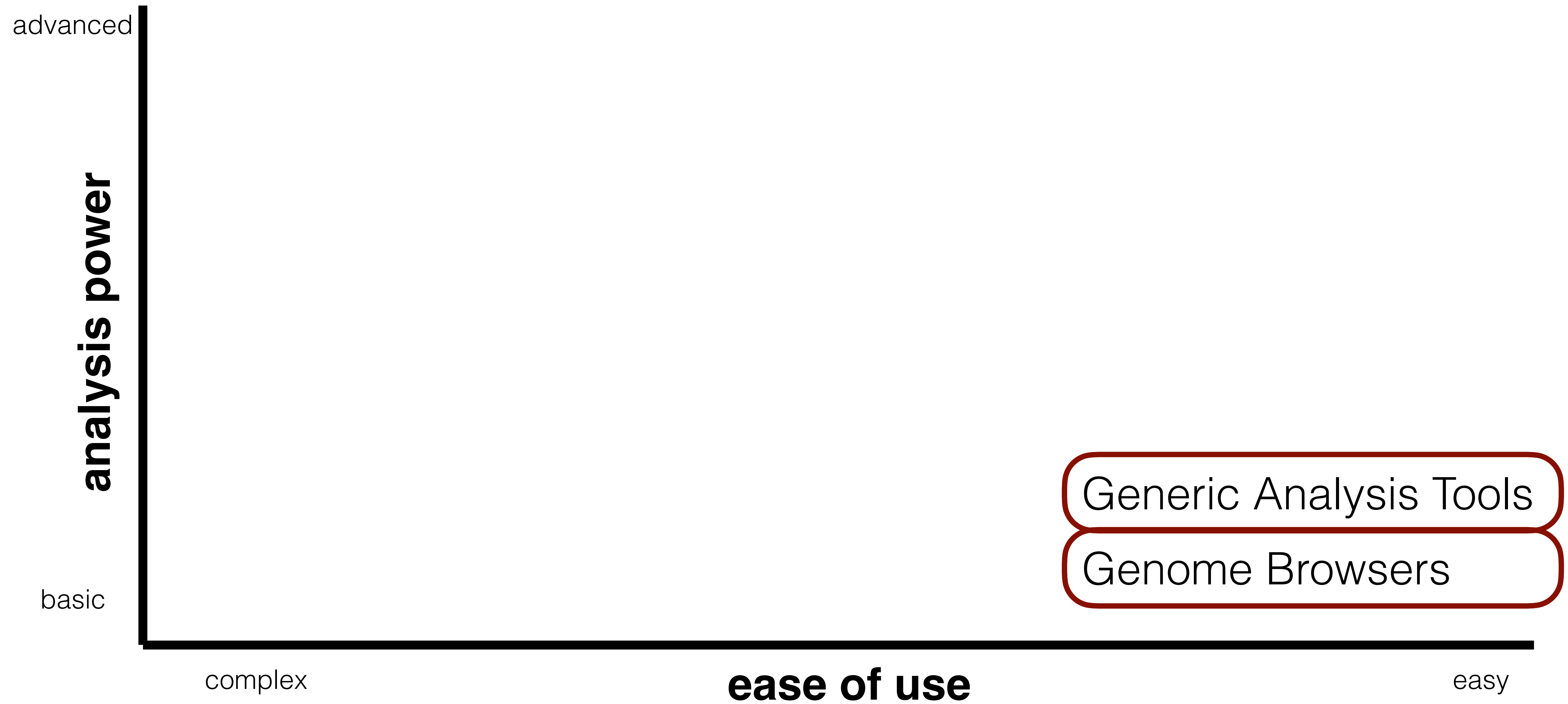
ease of use

easy

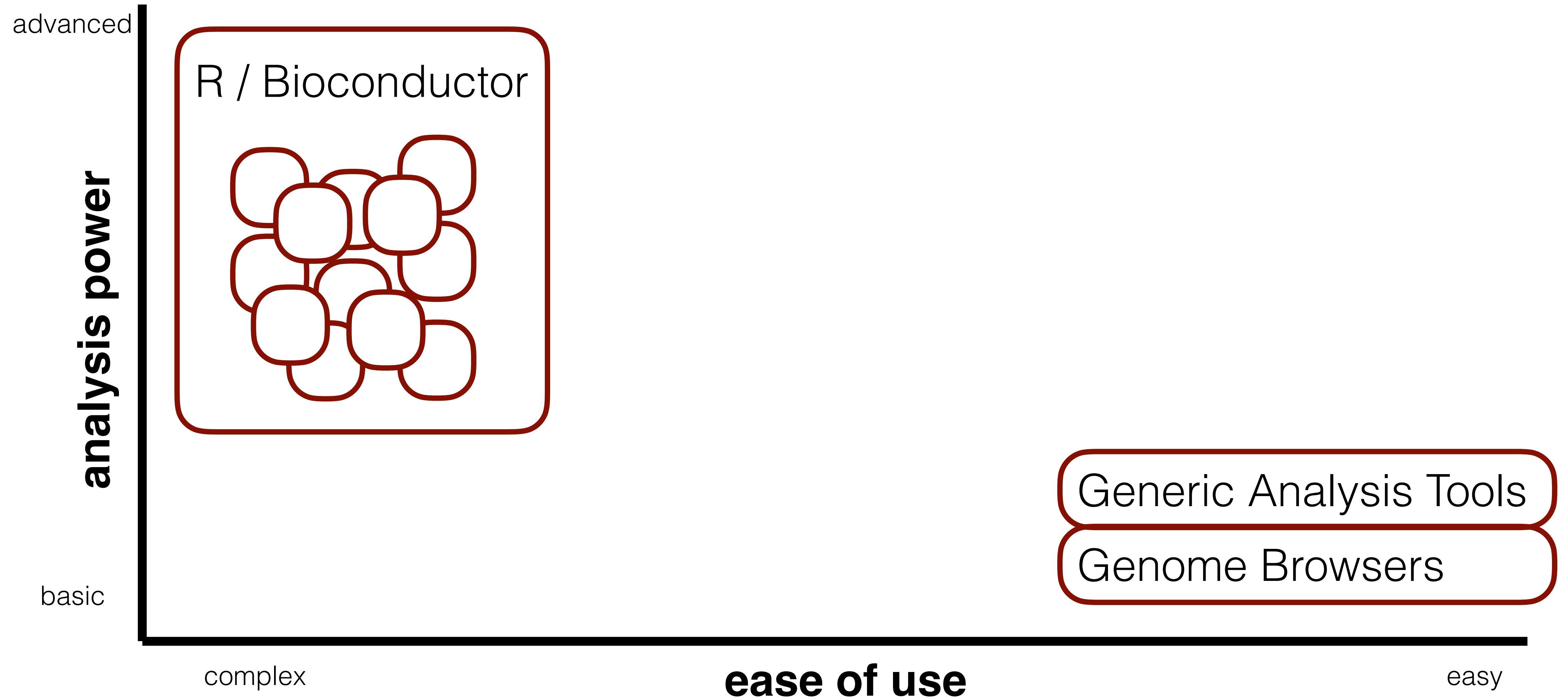
Generic Analysis Tools

Genome Browsers

motivation



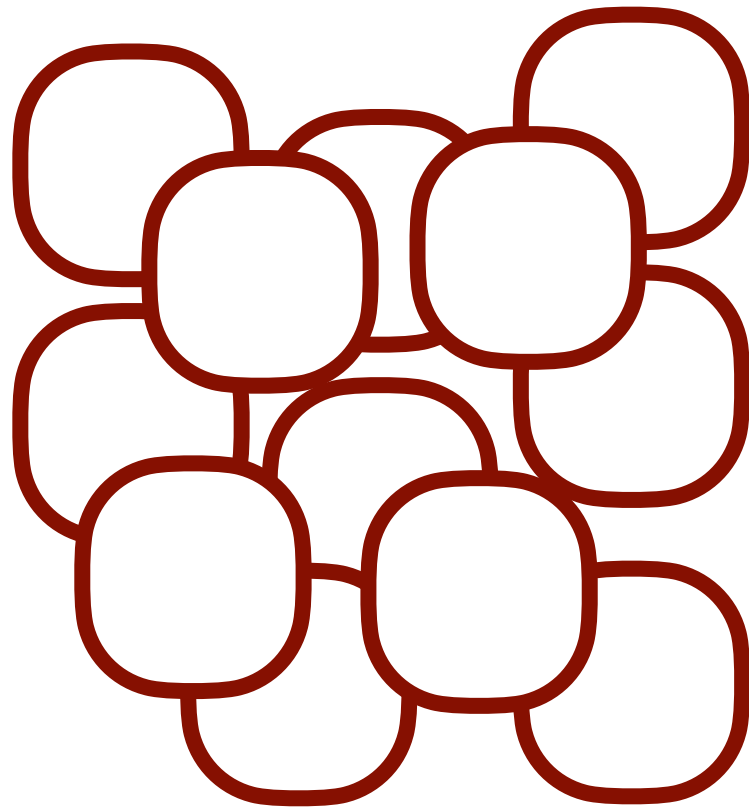
motivation



motivation

advanced

R / Bioconductor



analysis power

basic

complex

ease of use

easy

Packages found under Sequencing:

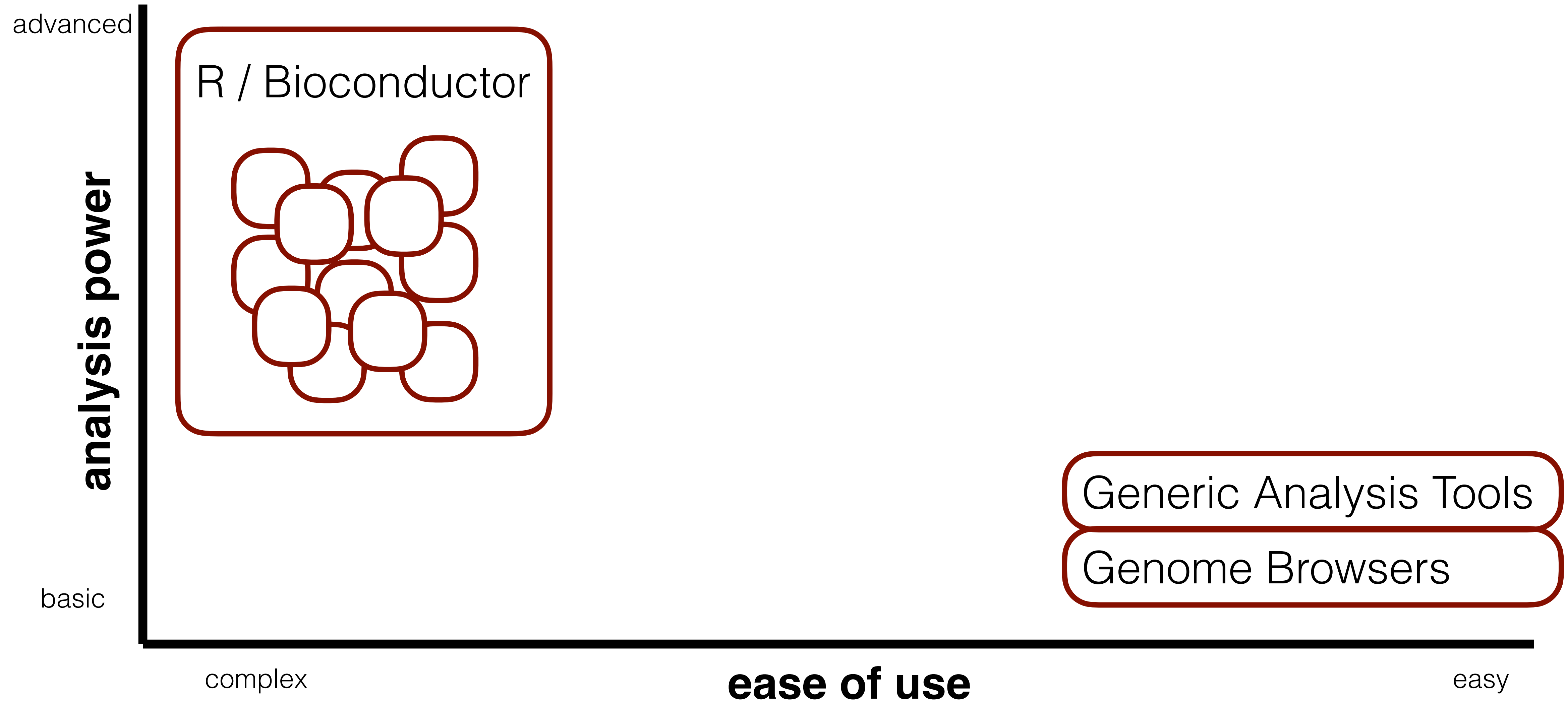
Package	Maintainer	Title
AIMS	Eric R Paquet	AIMS : Absolute Assignment of Breast Cancer Intrinsic Molecular Subtype
ALDEx2	Greg Gloor	Analysis of differential abundance taking sample variation into account
AllelicImbalance	Jesper R Gadin	Investigates allele specific expression
ampliQueso	Michal Okoniewski	Analysis of amplicon enrichment panels
AnnotationDbi	Bioconductor Package Maintainer	Annotation Database Interface
anota	Ola Larsson	ANalysis Of Translational Activity (ANOTA).
ArrayExpressHTS	Angela Goncalves, Andrew Tikhonov	ArrayExpress High Throughput Sequencing Processing Pipeline
BADER	Andreas Neudecker	Bayesian Analysis of Differential Expression in RNA Sequencing Data
ballgown	Alyssa Frazee	Flexible, isoform-level differential expression analysis
bamsignals	Alessandro Mammana	Extract read count signals from bam files

Bioconductor Repository

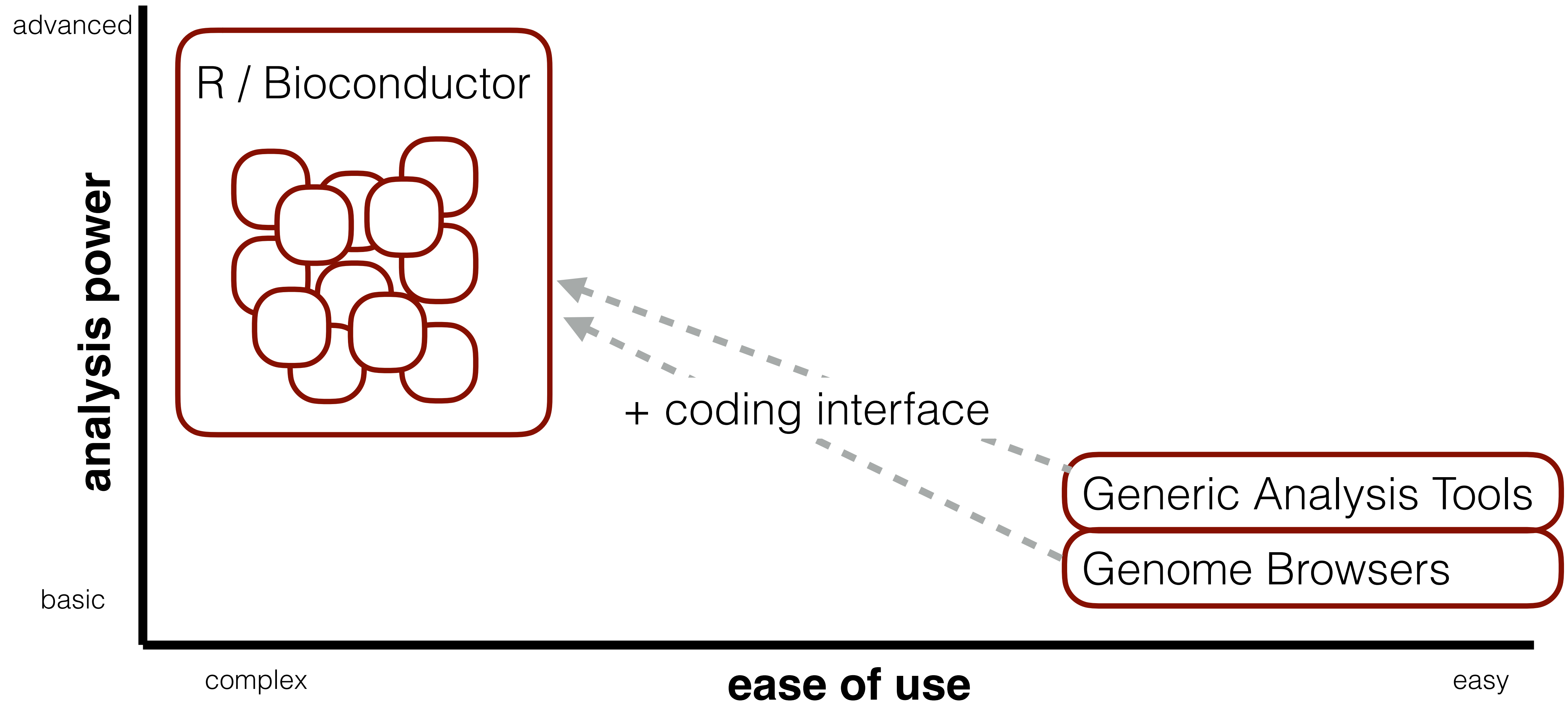
Generic Analysis Tools

Genome Browsers

motivation



motivation



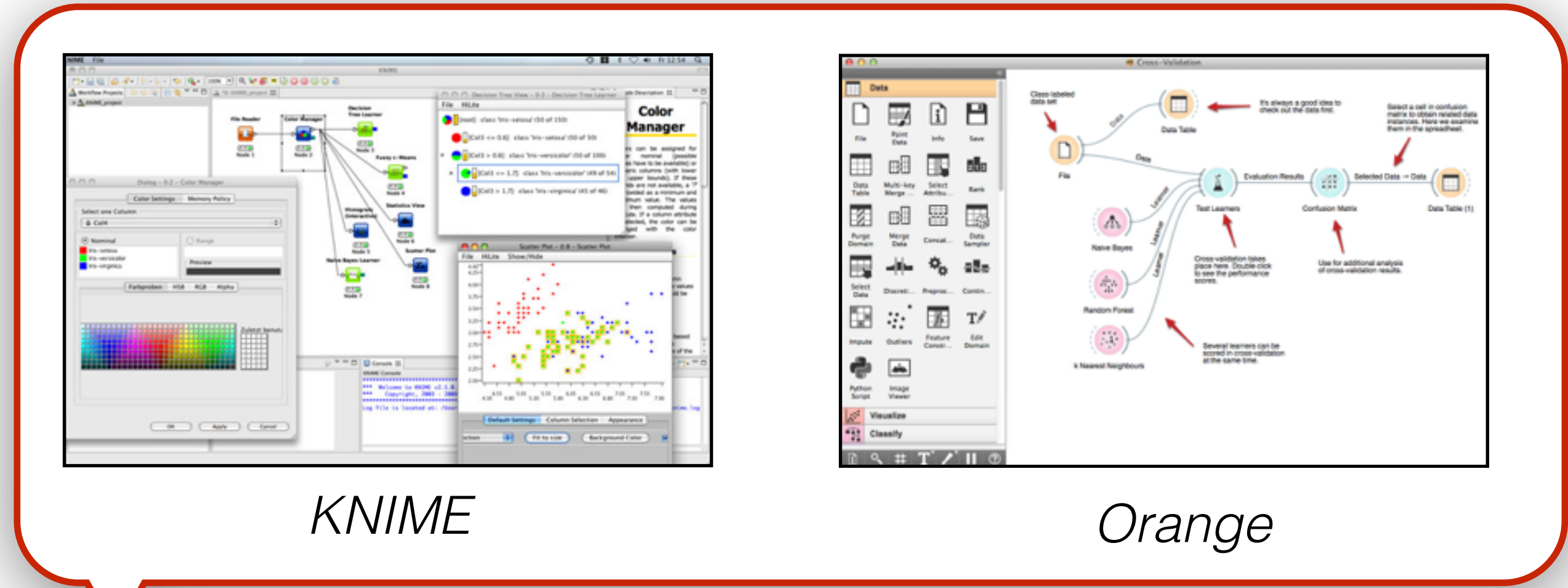
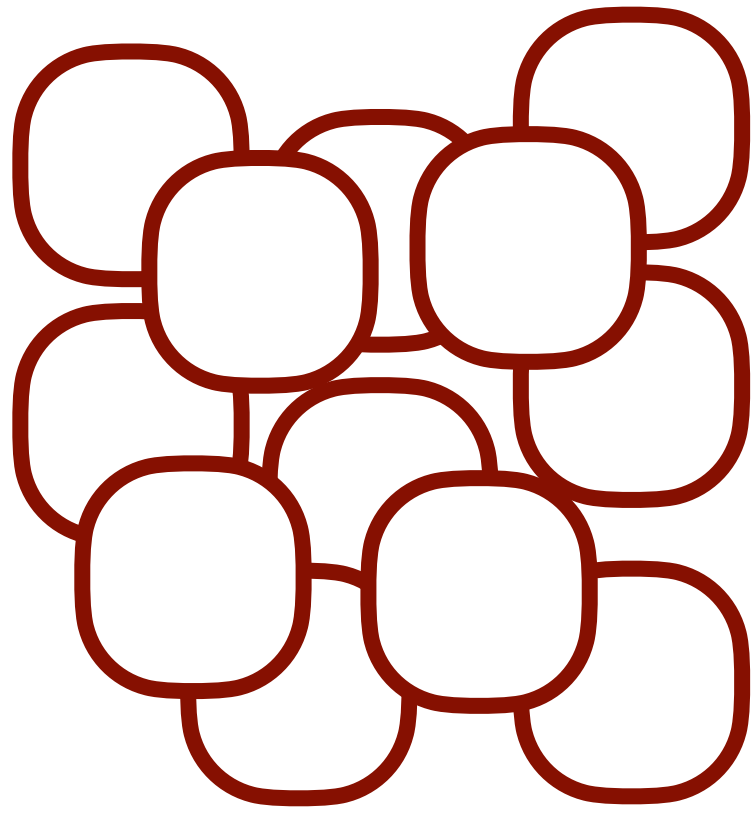
motivation

advanced

analysis power

basic

R / Bioconductor



KNIME

Orange

+ coding interface

Generic Analysis Tools

Genome Browsers

complex

ease of use

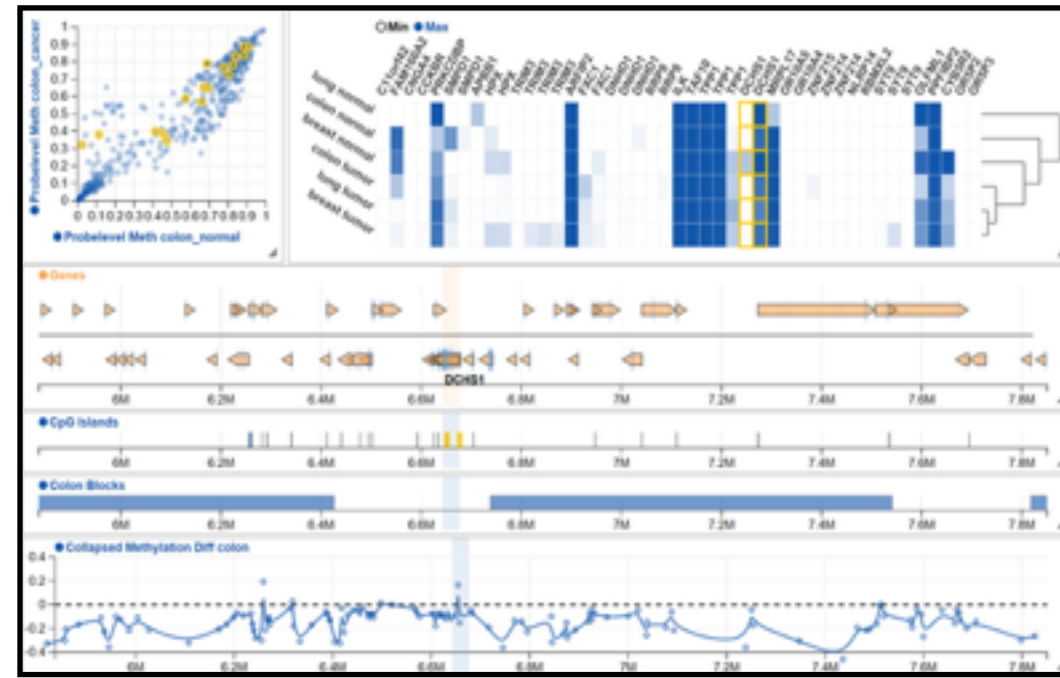
easy

motivation

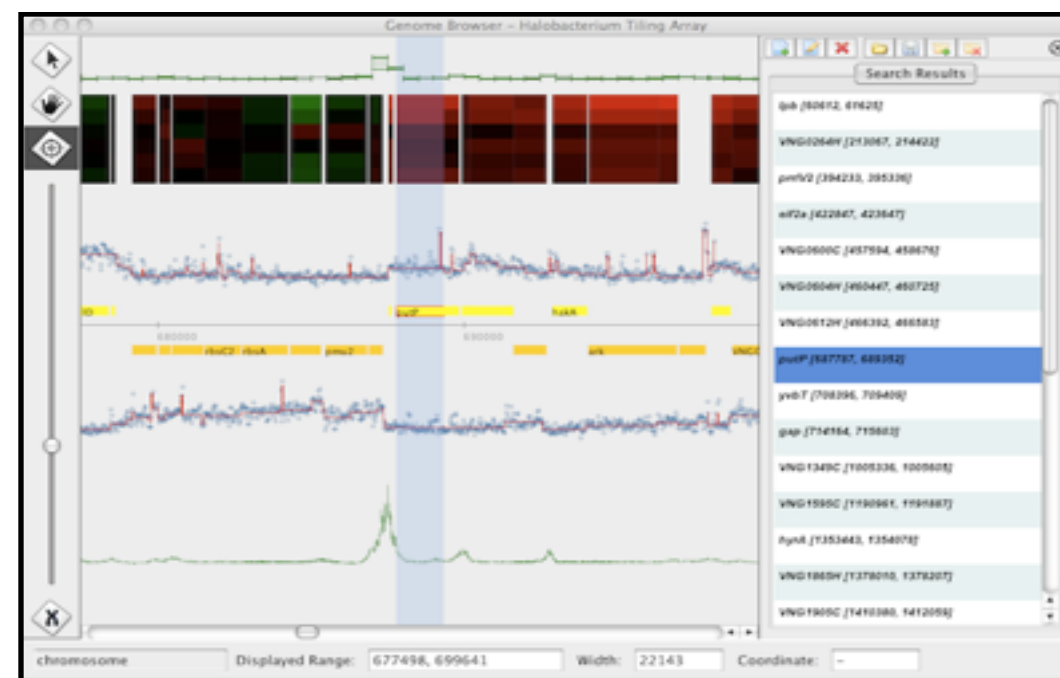
advanced

R / Bioconductor

analysis power

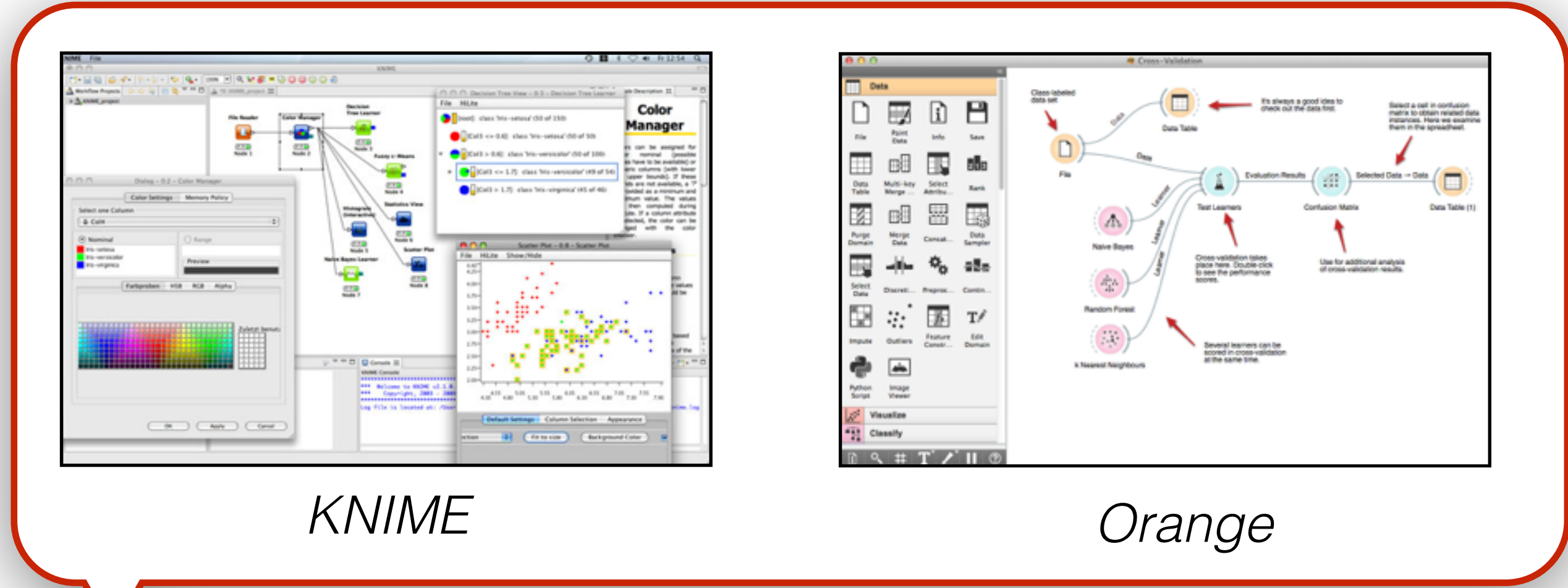


Epiviz



Gaggle Genome Browser

basic



KNIME

Orange

+ coding interface

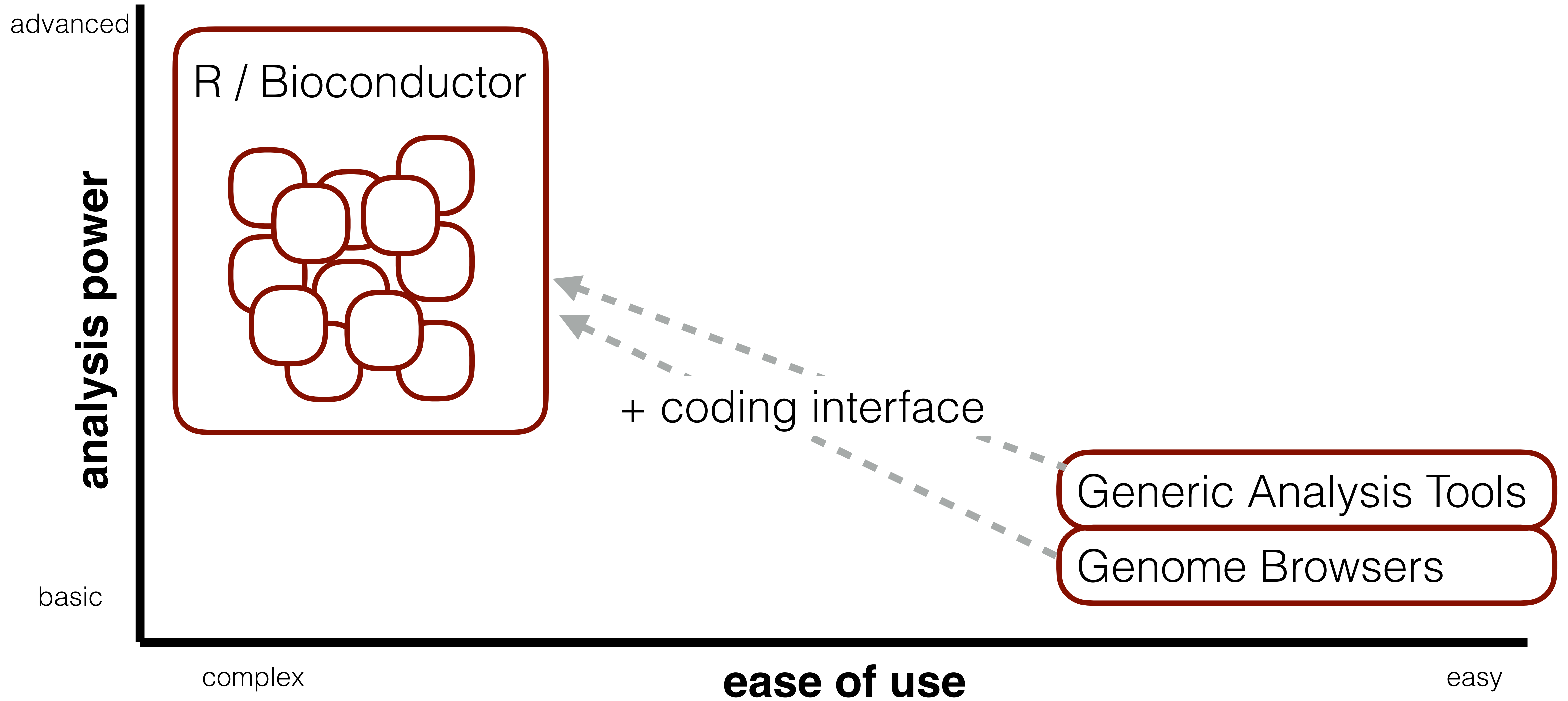
Generic Analysis Tools

Genome Browsers

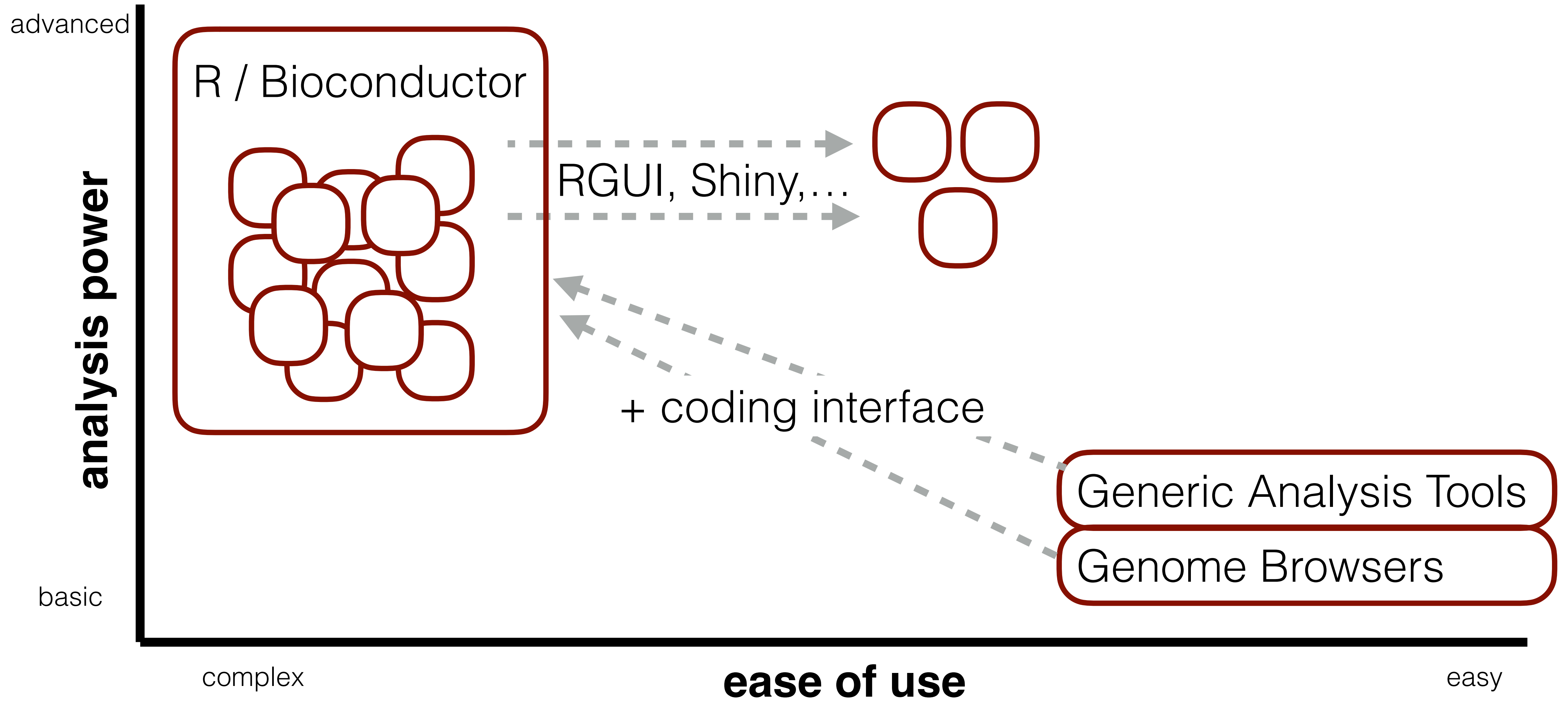
ease of use

easy

motivation



motivation



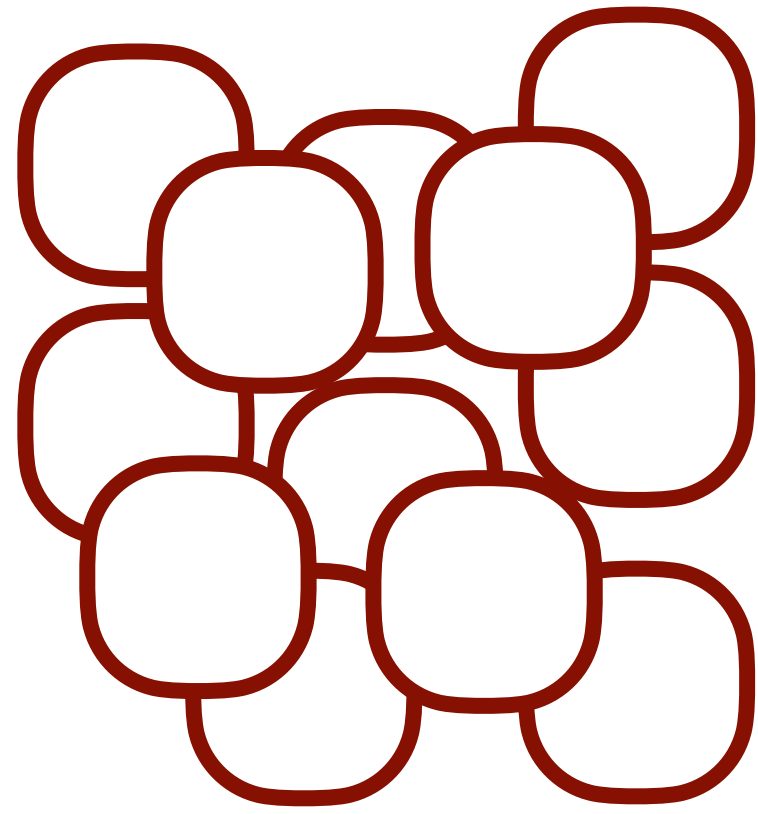
motivation

advanced

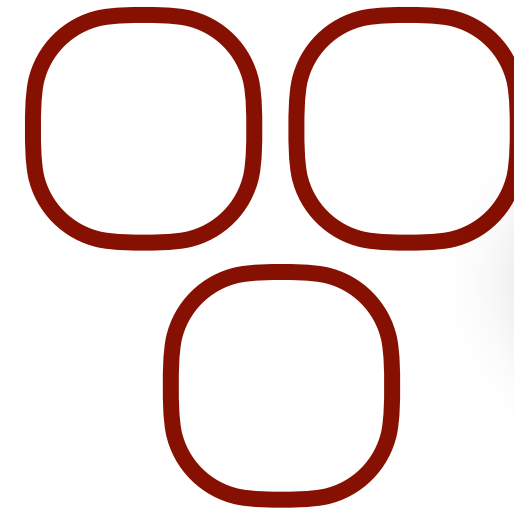
analysis power

basic

R / Bioconductor



RGUI, Shiny, ...

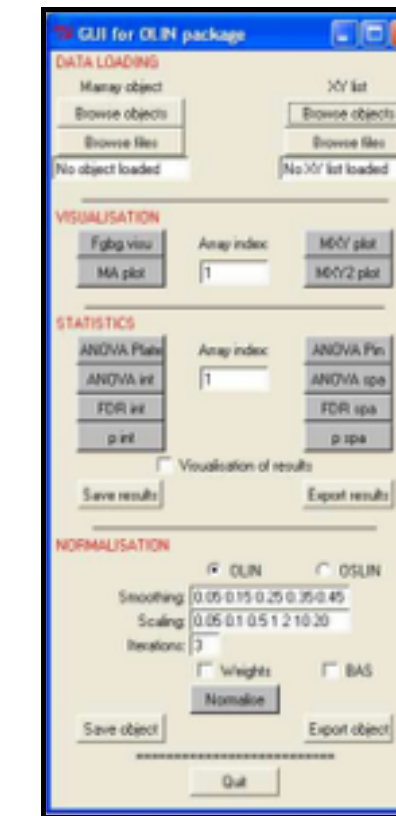


+ coding interface

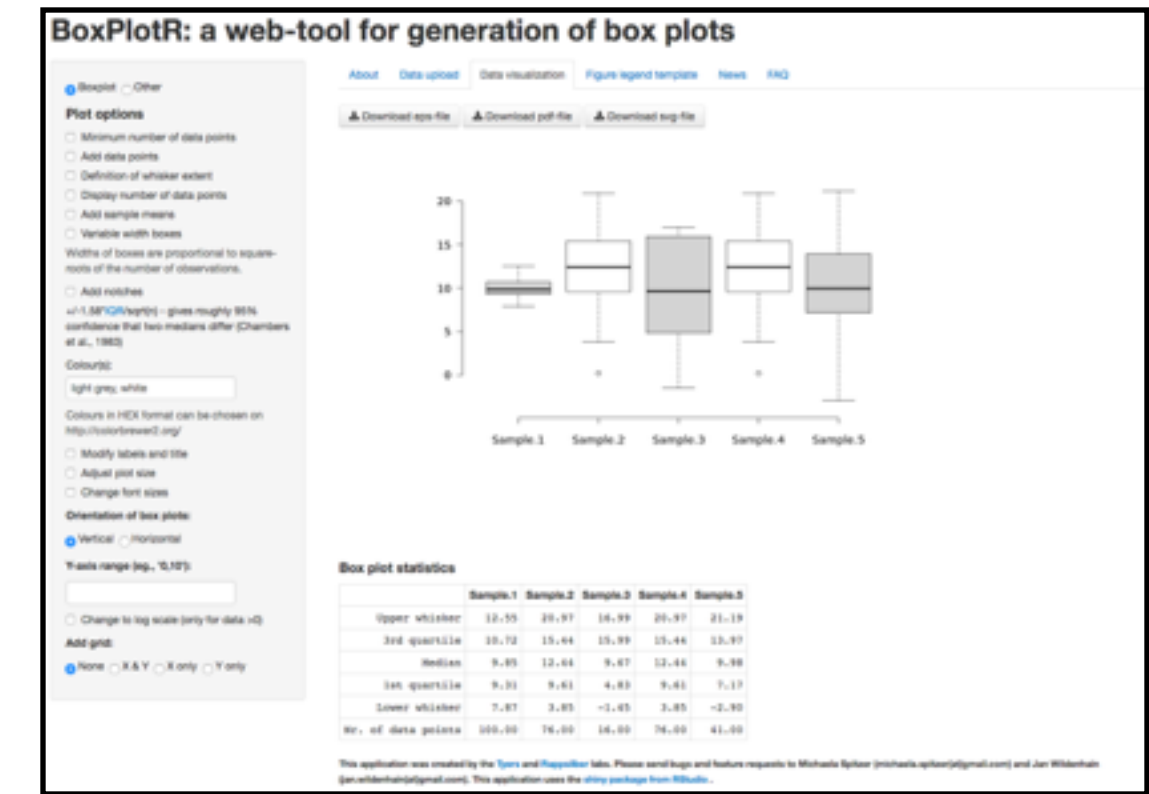
complex

ease of use

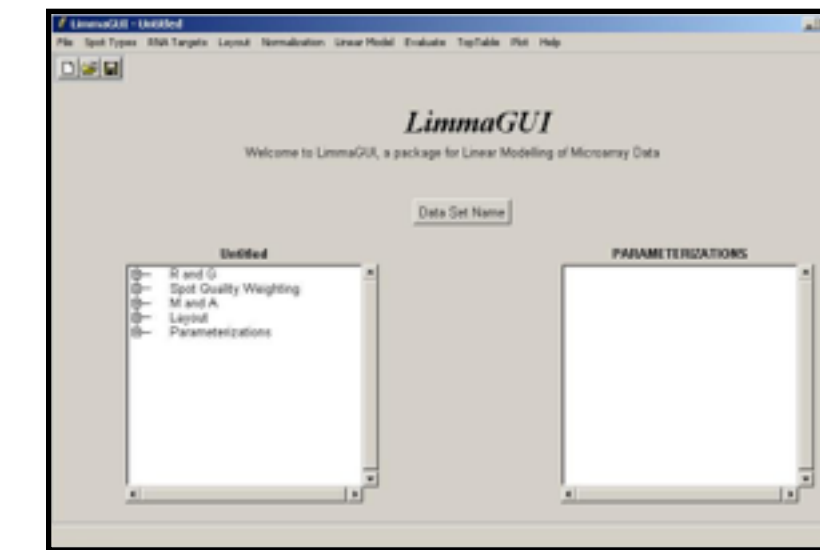
easy



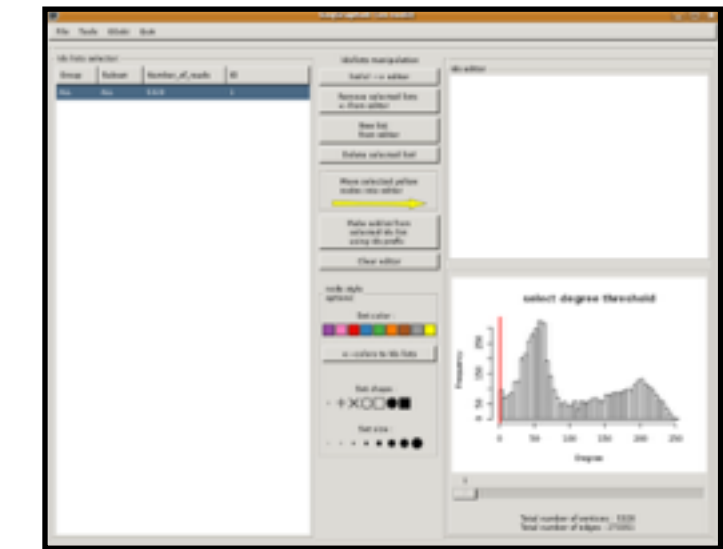
OLINGui



BoxPlotR



LimmaGUI

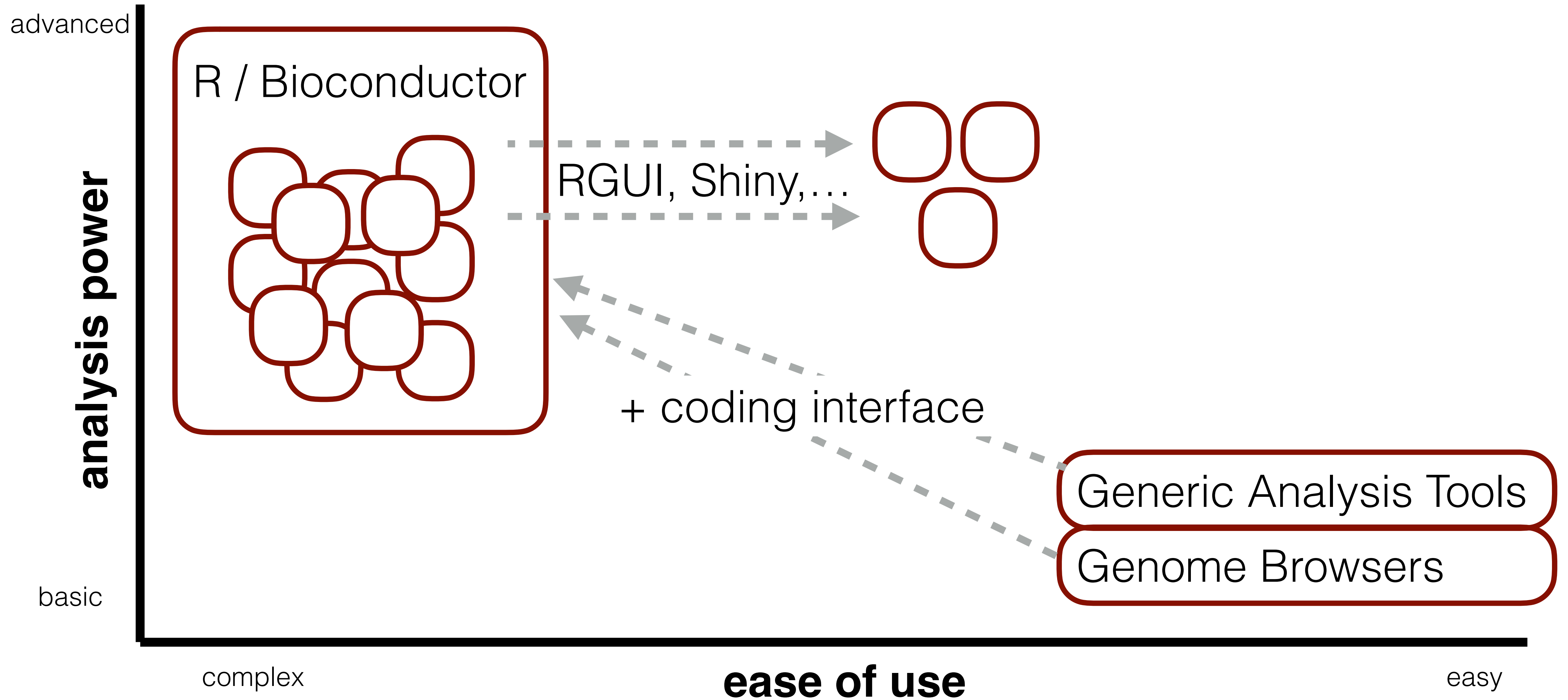


SeqGraphR

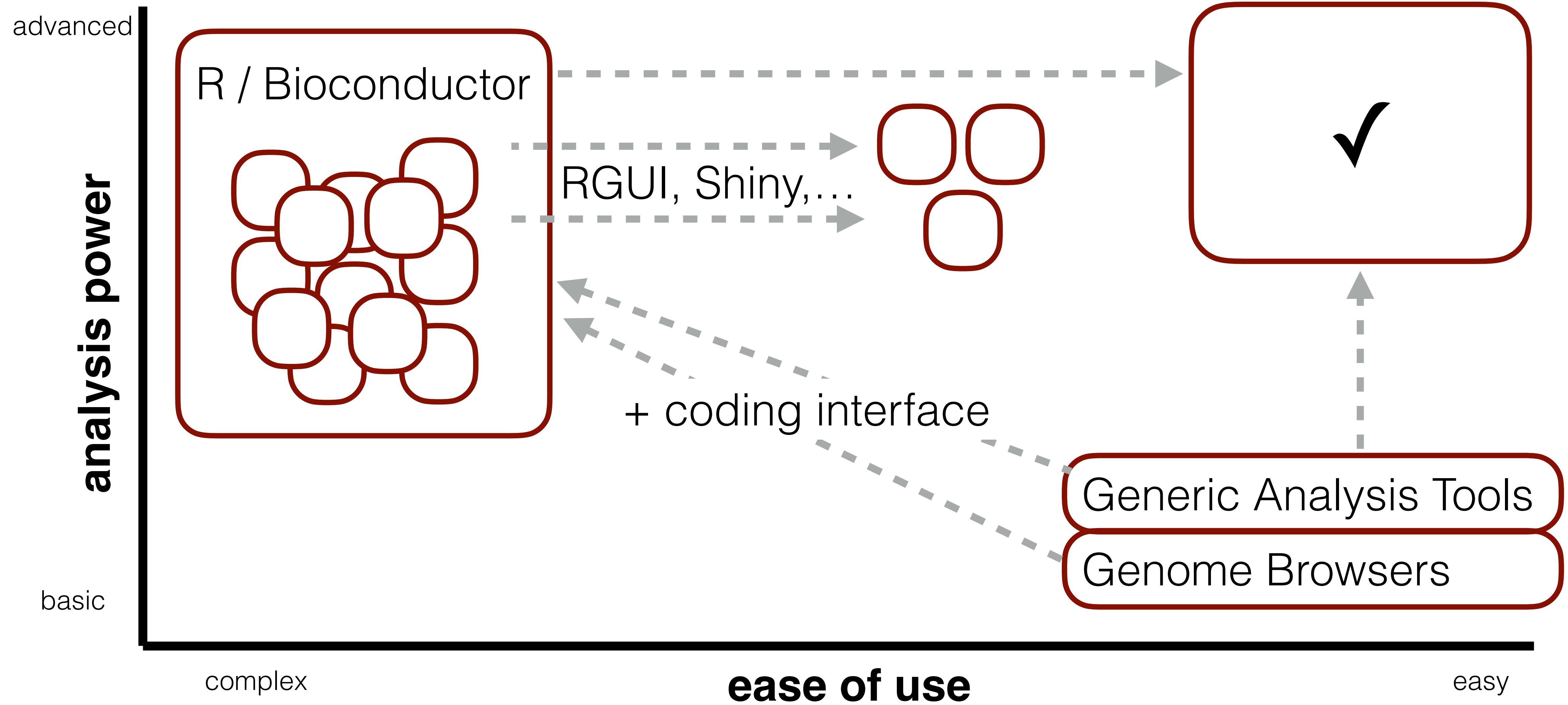
Generic Analysis Tools

Genome Browsers

motivation

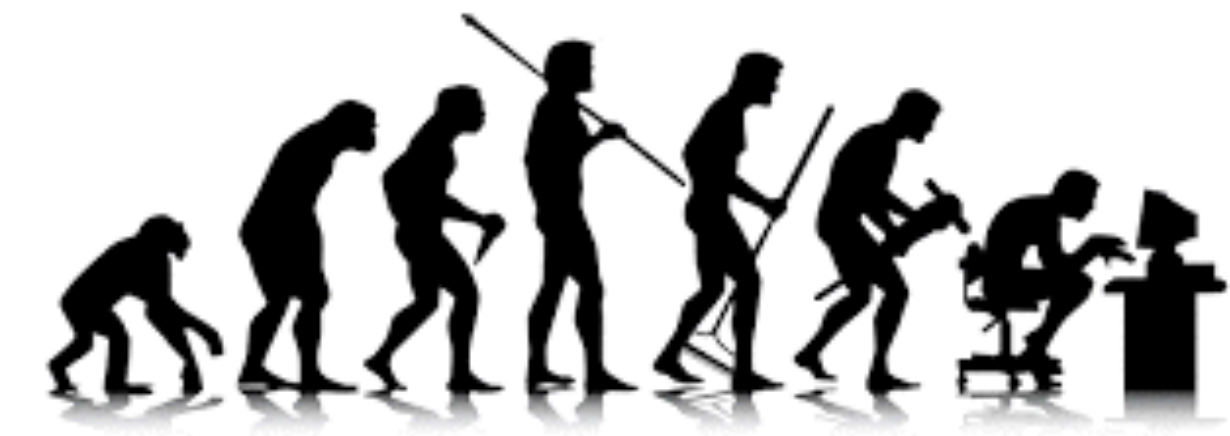


motivation



VisRseq

design evolution



IGV plugin
(2013)

workspace

R
integration

BioVis Poster
(2014)

Redesigned
Layout

BMC Bioinf
(2015)

open beta



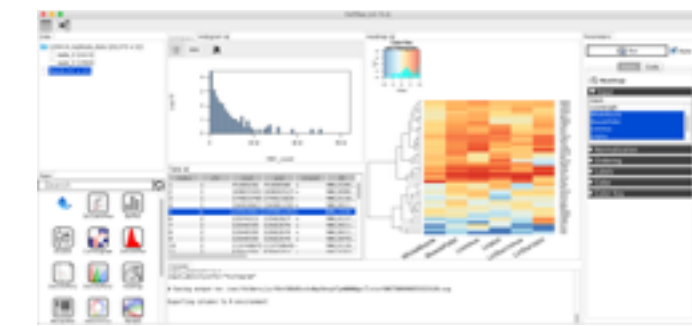
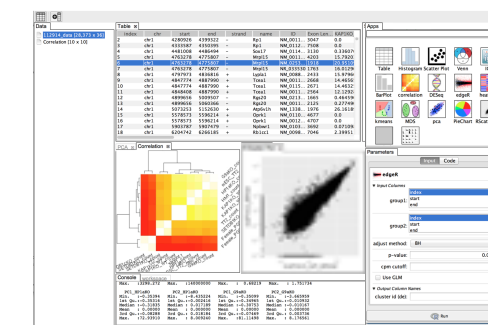
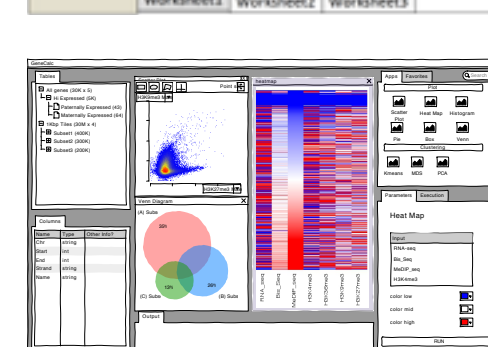
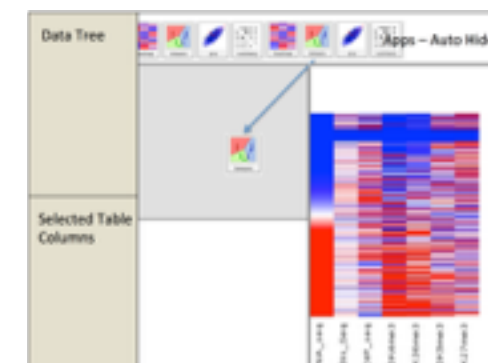
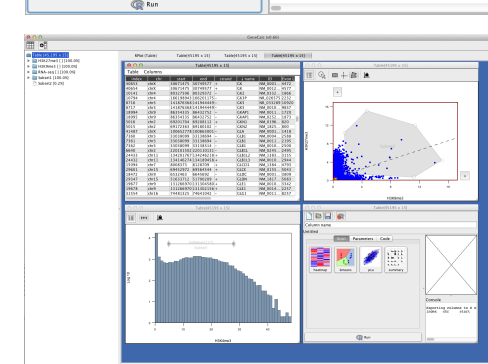
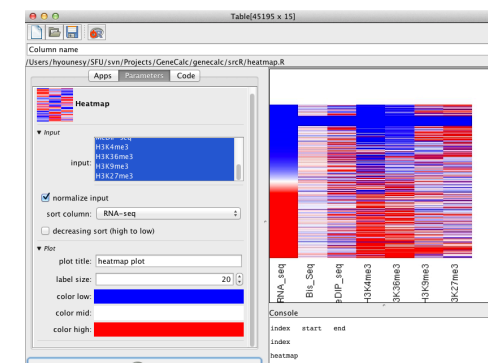
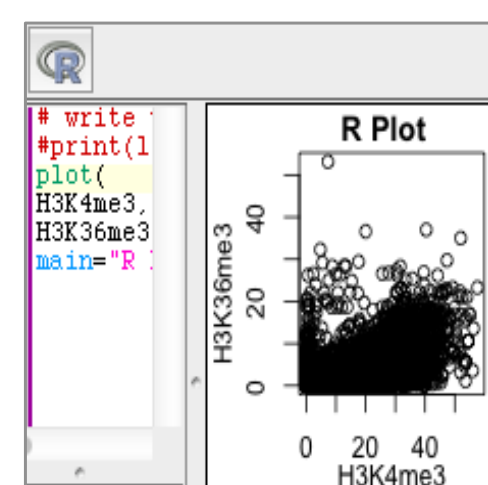
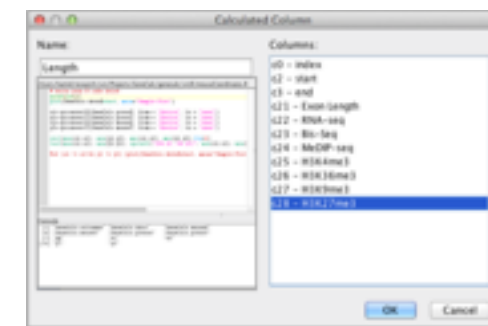
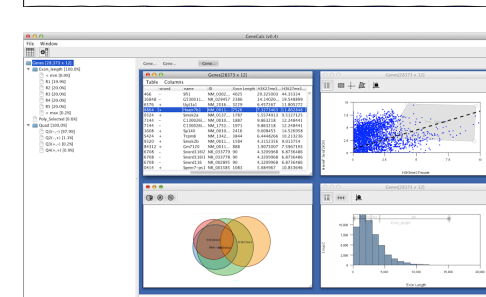
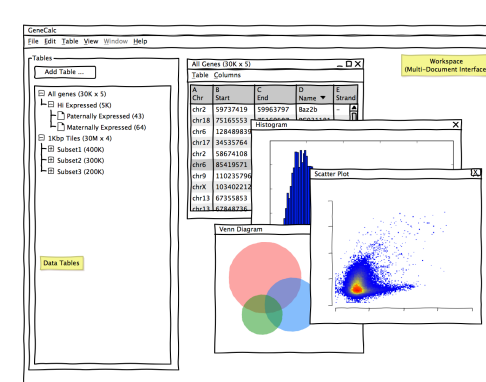
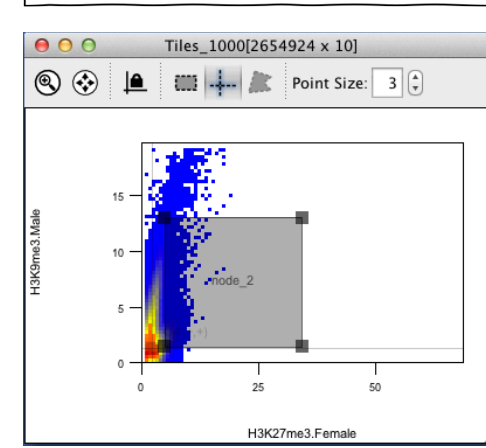
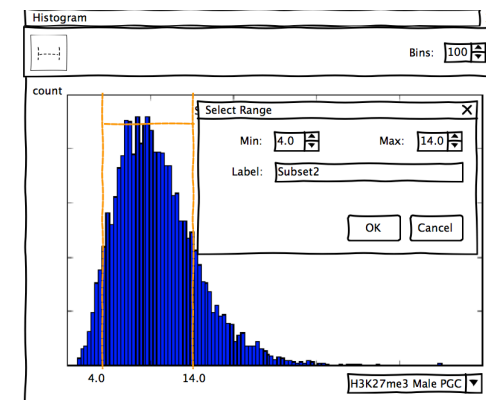
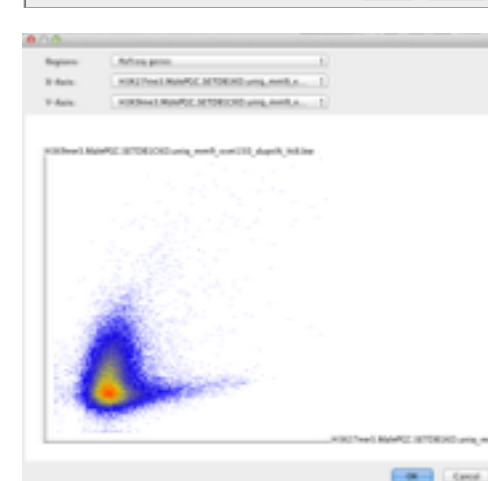
Add Expression Column

Expression: C = B
Column Name: Length

B	Start	7	8	9	/	ABS
C	End	4	5	6	x	
F	H3K9me3	1	2	3	-	
G	H3K27me3	0	()	+	

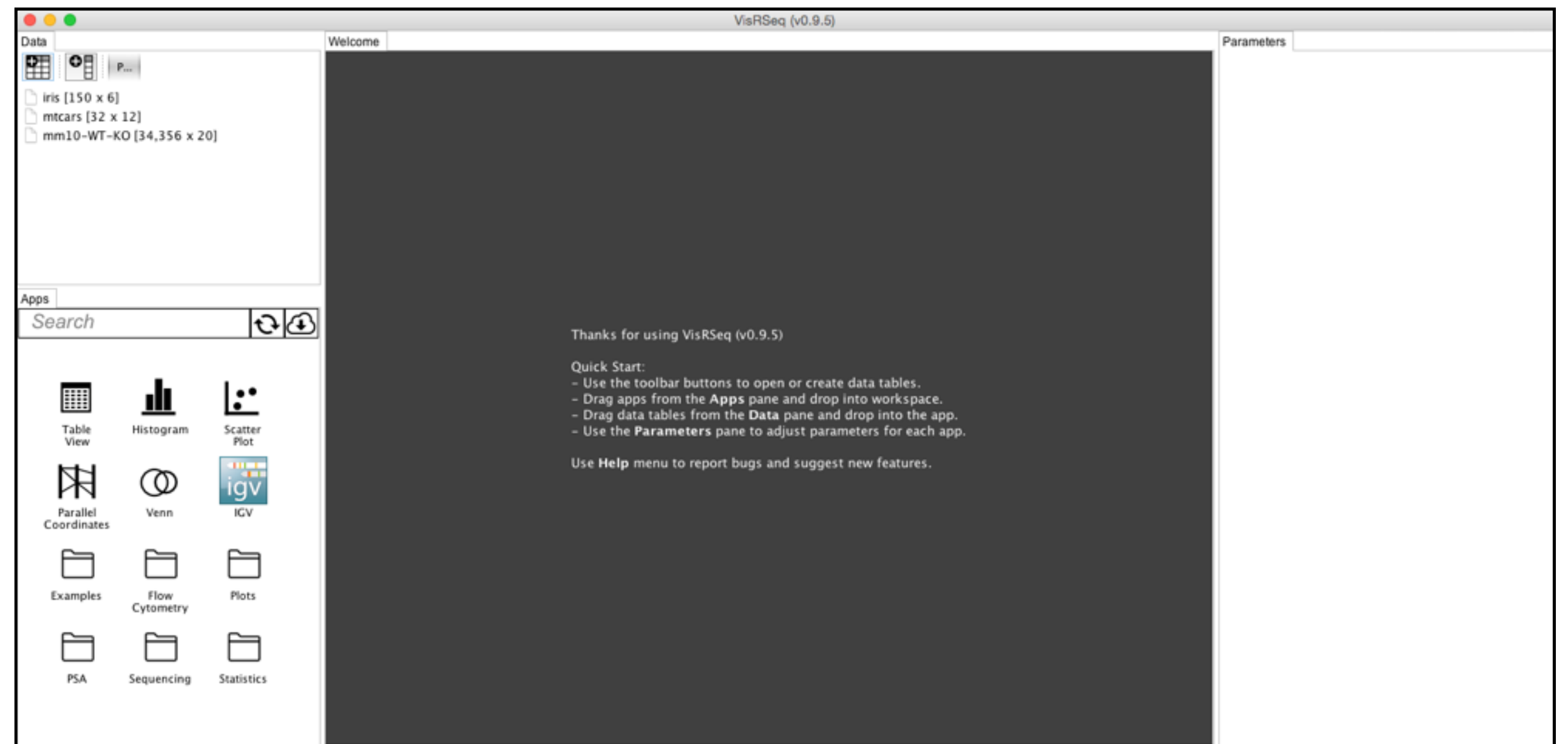
Calculated Columns

Name	Columns
Length	C2 - index
	C2 - start
	C2 - end
	C23 - Exon Length
	C22 - RNA-seq
	C23 - Bis-Seq
	C24 - MeDP-seq
	C25 - H3K4me3
	C26 - H3K36me3
	C27 - H3K9me3
	C28 - H3K27me3

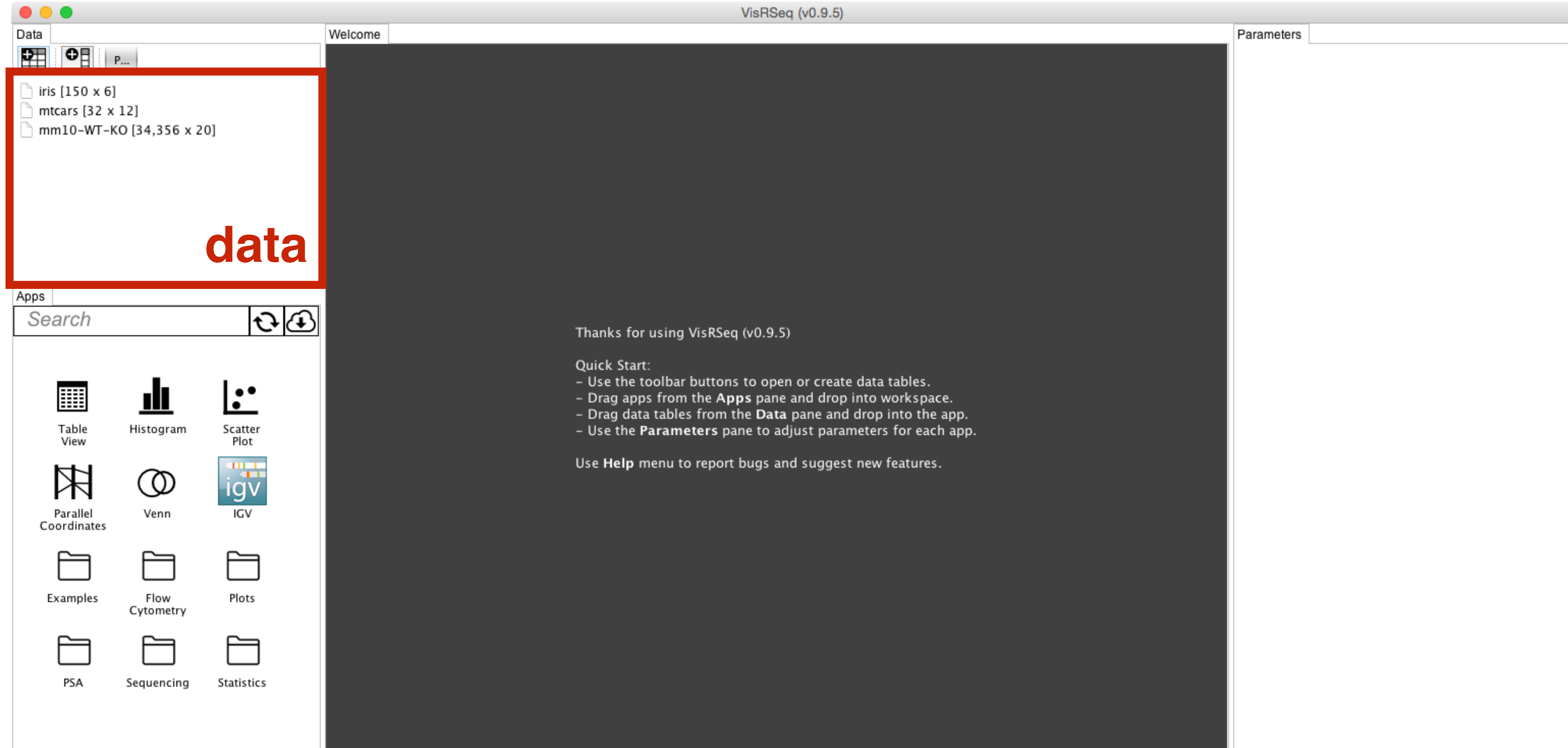


interface

- Installation Requirements:
 - Java SE 7
 - R
- Auto installs required packages
- Open Beta: <http://visrseq.github.io>



interface: data



interface: data

Tab delimited Text

The screenshot displays a data analysis interface with a spreadsheet window titled 'iris.txt' showing the following data:

	A	B	C	D	E
1	Sepal.Length	Sepal.Width	Petal.Length	Petal.Width	Species
2	5.1	3.5	1.4	0.2	setosa
3	4.9	3	1.4	0.2	setosa
4	4.7	3.2	1.3	0.2	setosa
5	4.6	3.1	1.5	0.2	setosa
6	5	3.6	1.4	0.2	setosa
7	5.4	3.9	1.7	0.4	setosa
8	4.6	3.4	1.4	0.3	setosa
9	5	3.4	1.5	0.2	setosa
10	4.4	2.9	1.4	0.2	setosa
11	4.9	3.1	1.5	0.1	setosa
12	5.4	3.7	1.5	0.2	setosa
13	4.8	3.4	1.6	0.2	setosa
14	4.8	3	1.4	0.1	setosa

A 'Calculated Column' dialog box is open, showing the following details:

- New Column Name: zscore
- Equation = ZSCORE(c26, c27)
- Numerical Columns: c27 - RNA-seq Male, c28 - H3K36me3_CASTEij, c29 - H3K36me3_CS7BL6J, c30 - RNA-Seq_CASTEij_RPKM, c31 - RNA-Seq_CS7BL6J_RPKM, c22 - H3K26me2_CASTEij_RPKM
- Function list: ABS, SQRT, LN, LOG, LOG10, EXP, POWER, INT, FLOOR, CEILING, MOD, MIN, MAX, AVERAGE, STDEV, MEDIAN, SUM, COUNT, ZSCORE, IF, NOT, AND, OR

The background interface includes a 'Data' pane with a list of datasets (iris [150 x 6], mtcars [32 x 12], mm10-WT-KO [34,35]), an 'Apps' pane with a search bar and icons for Table View, Histogram, Parallel Coordinates, Venn, IGV, Flow Cytometry, and Sequencing, and a 'Parameters' pane.

interface: data

Tab delimited Text

.BAM, .WIG

The screenshot shows an Excel spreadsheet with the following data:

	A	B	C	D	E
1	Sepal.Length	Sepal.Width	Petal.Length	Petal.Width	Species
2	5.1	3.5	1.4	0.2	setosa
3	4.9	3	1.4	0.2	setosa
4	4.7	3.2	1.3	0.2	setosa
5	4.6	3.1	1.5	0.2	setosa
6	5	3.6	1.4	0.2	setosa
7	5.4	3.9	1.7	0.4	setosa
8	4.6	3.4	1.4	0.3	setosa
9	5	3.4	1.5	0.2	setosa
10	4.4	2.9	1.4	0.2	setosa
11	4.9	3.1	1.5	0.1	setosa
12	5.4	3.7	1.5	0.2	setosa
13	4.8	3.4	1.6	0.2	setosa
14	4.8	3	1.4	0.1	setosa

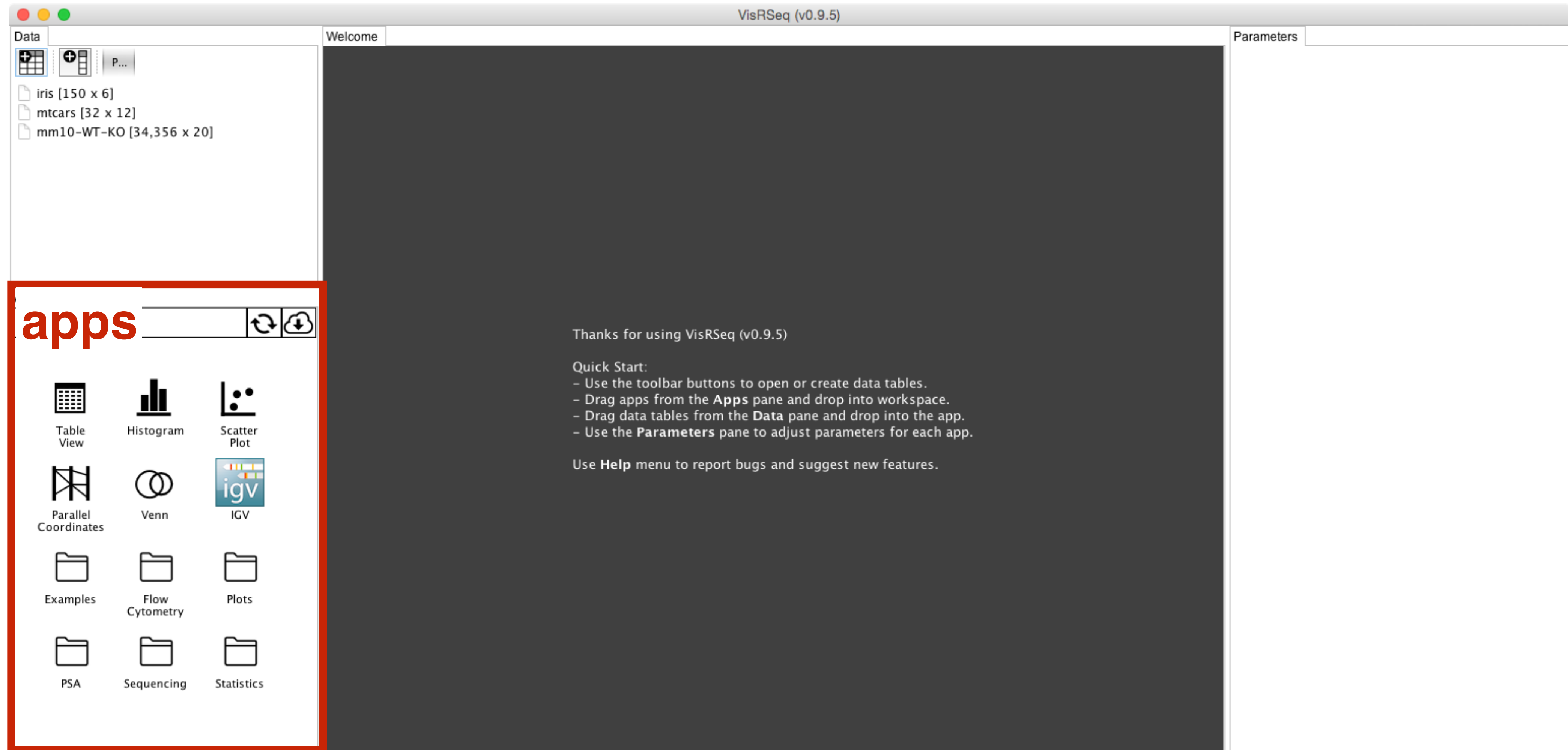
The 'Add Data Column' dialog box in IGV shows the following configuration:

- Select Data:** Rat_Oocyte_2_S4_001.bam (BAM), Rat_Oocyte_1_S3_001.bam (BAM)
- Column Name:** (empty)
- Regions:** Range selected. Start: Region Start, End: Region End, Number of Bins: 1
- Alignment Track Options:** Remove Duplicate Reads checked, Minimum Read Quality: 1
- Output:** RPKM checked, Read Count checked, Normalized by: total reads, Tracks: Gene, Read Length for BW tracks: 100

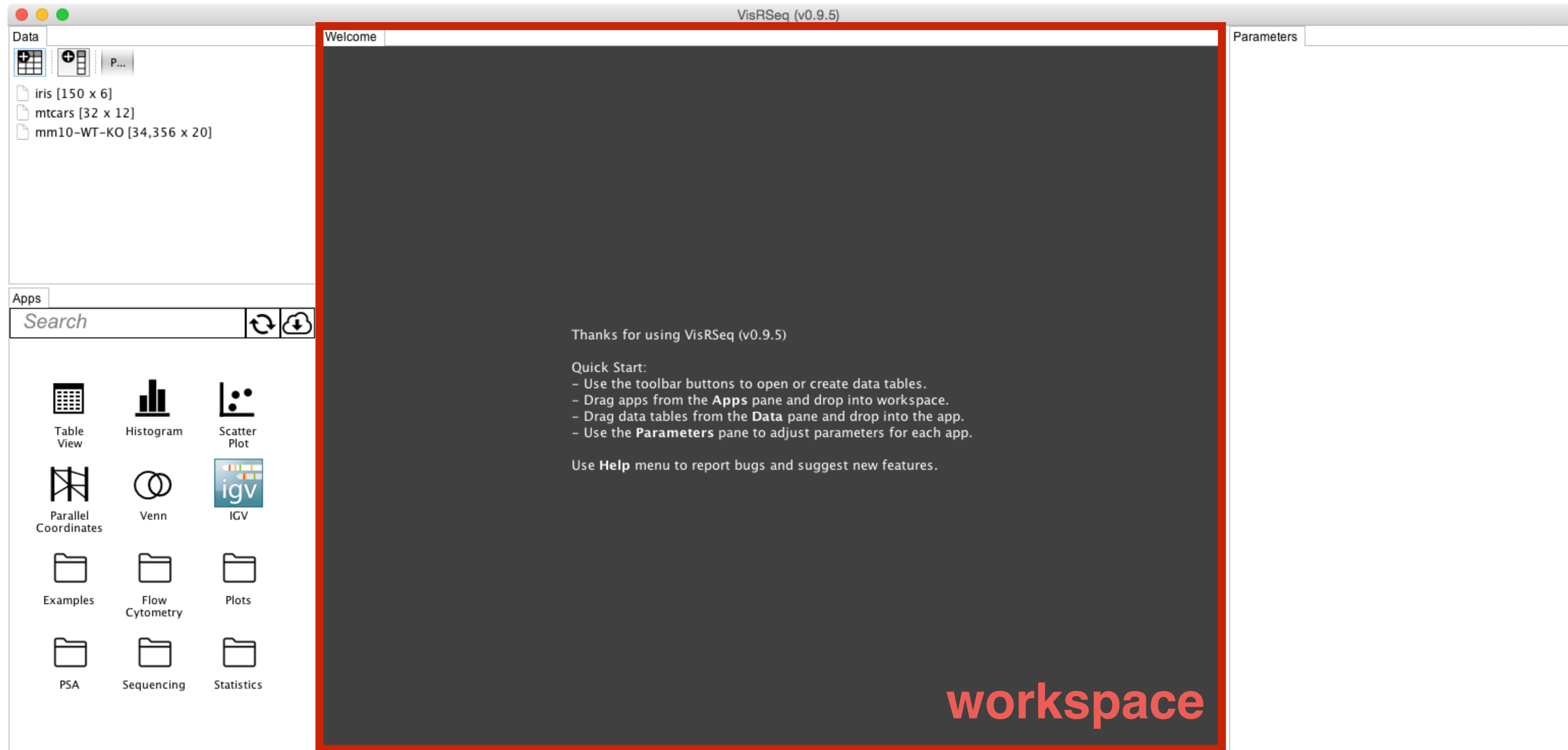
The 'Calculated Column' dialog box in IGV shows the following configuration:

- New Column Name:** zscore
- Equation =** ZSCORE(c26, c27)
- Numerical Columns:** c27 - RNA-seq Male, c28 - H3K36me3_CASTEij, c29 - H3K36me3_CS7BL6J, c30 - RNA-Seq_CASTEij_RPKM, c31 - RNA-Seq_CS7BL6J_RPKM, c22 - H3K26me3_CASTEij_RPKM

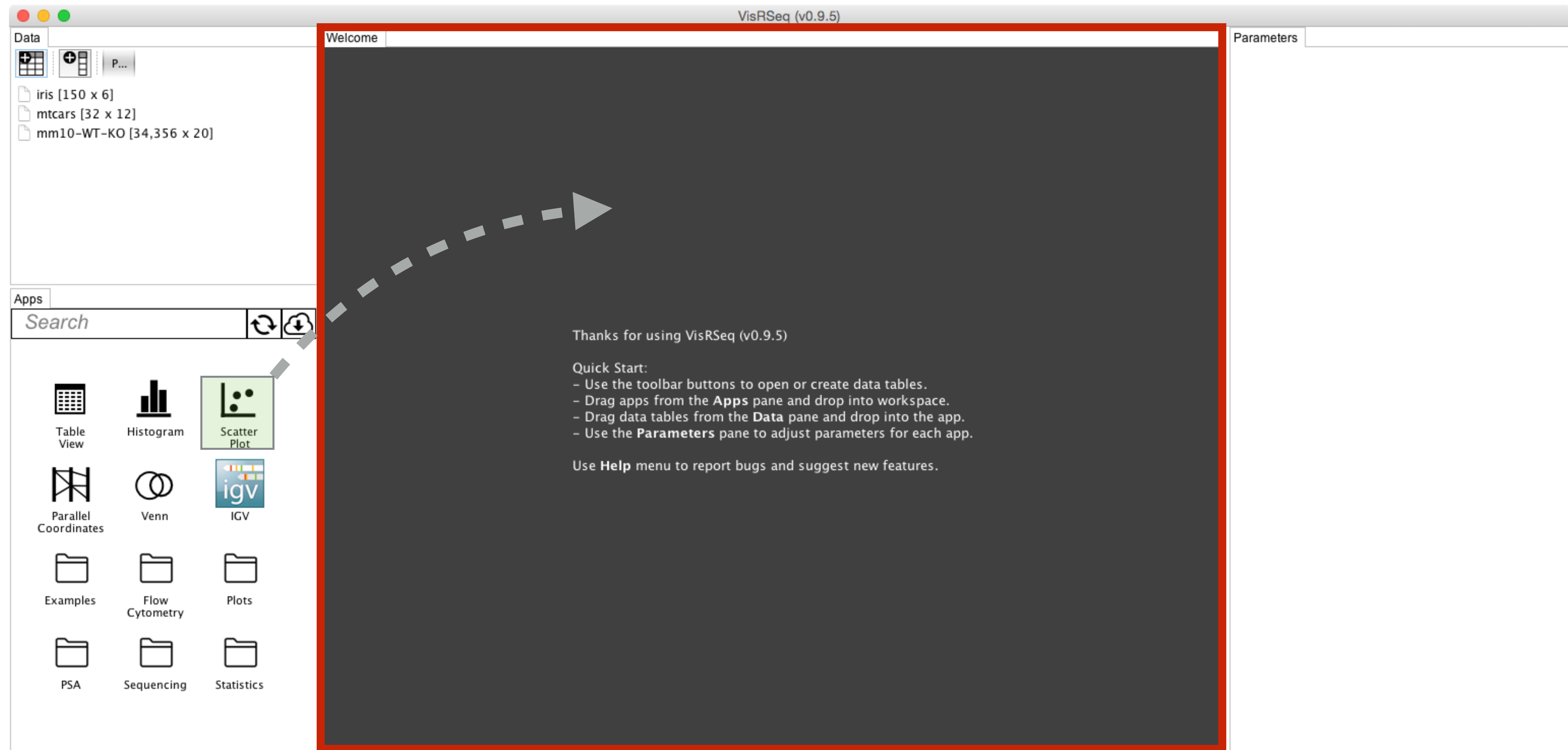
interface: apps



interface: workspace



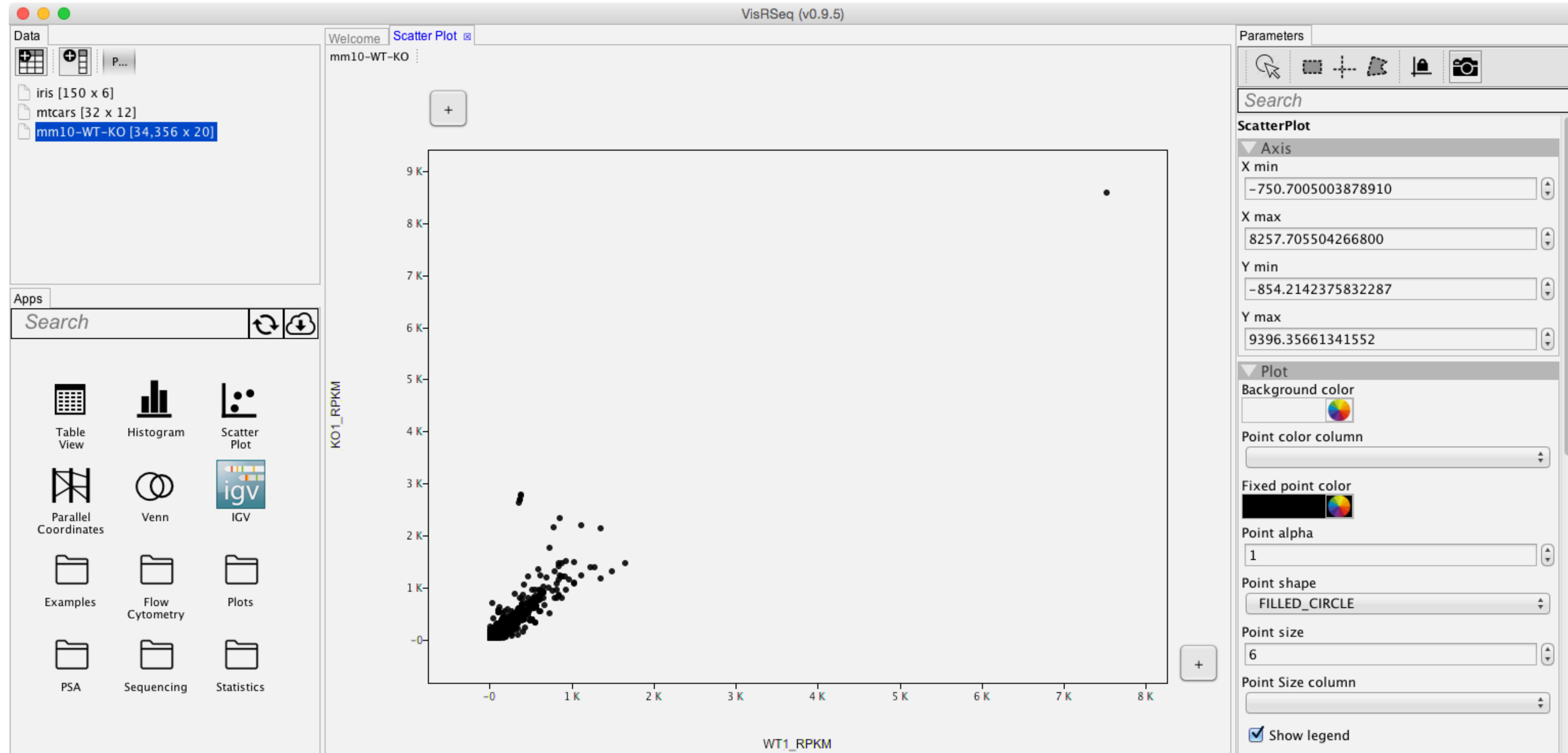
interface: workspace



interface: workspace

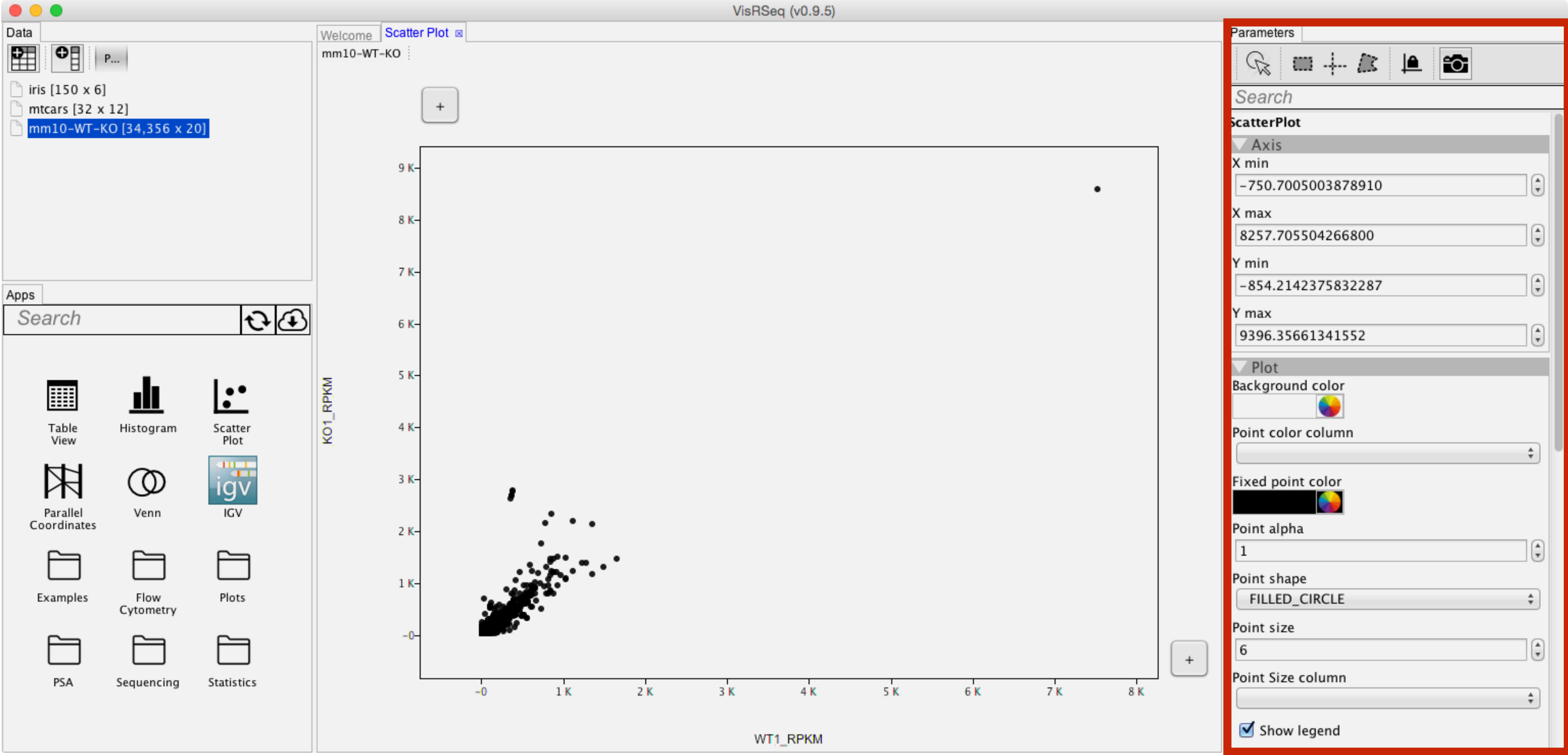


interface: workspace



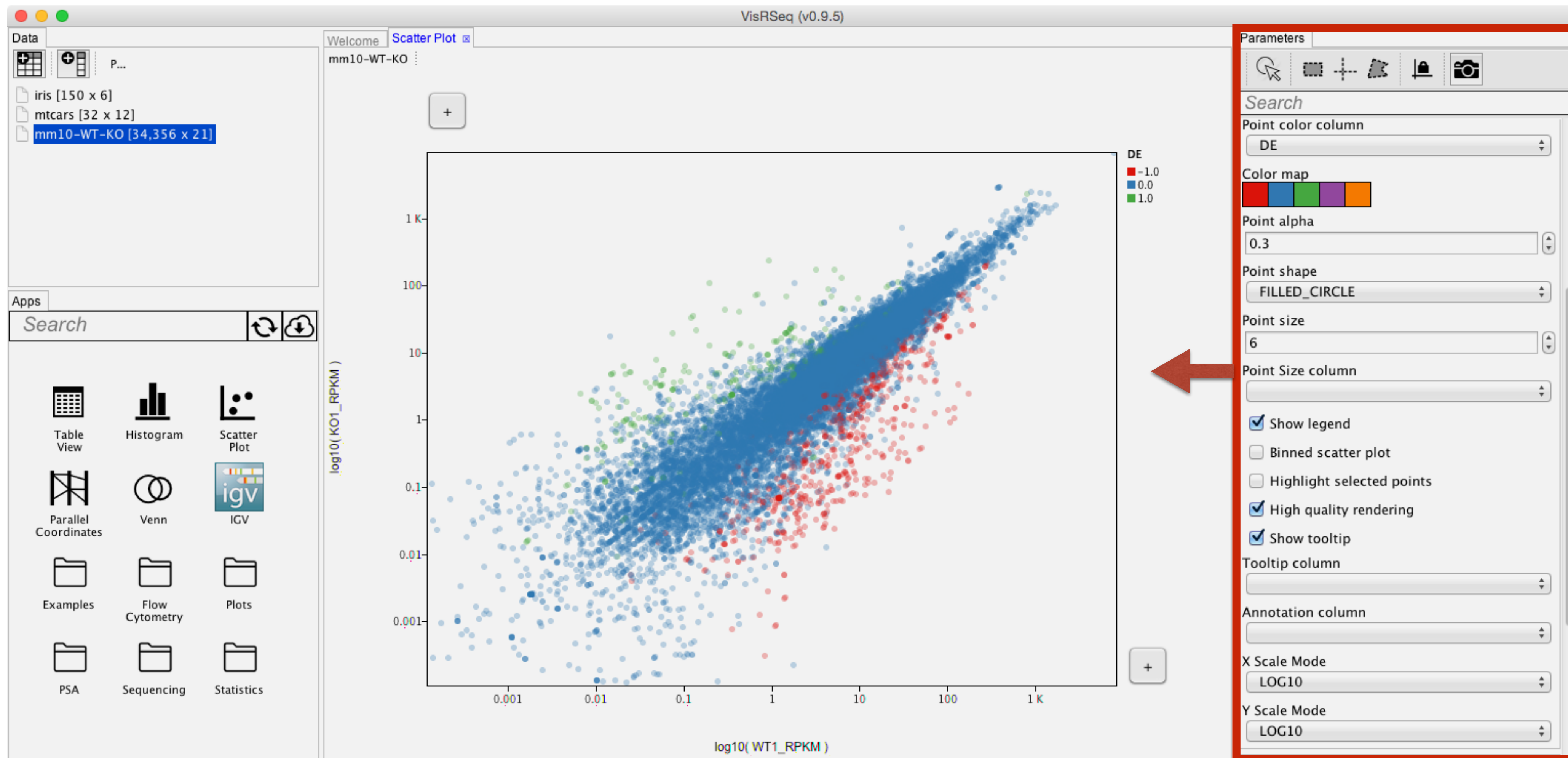
interface: parameters

parameters

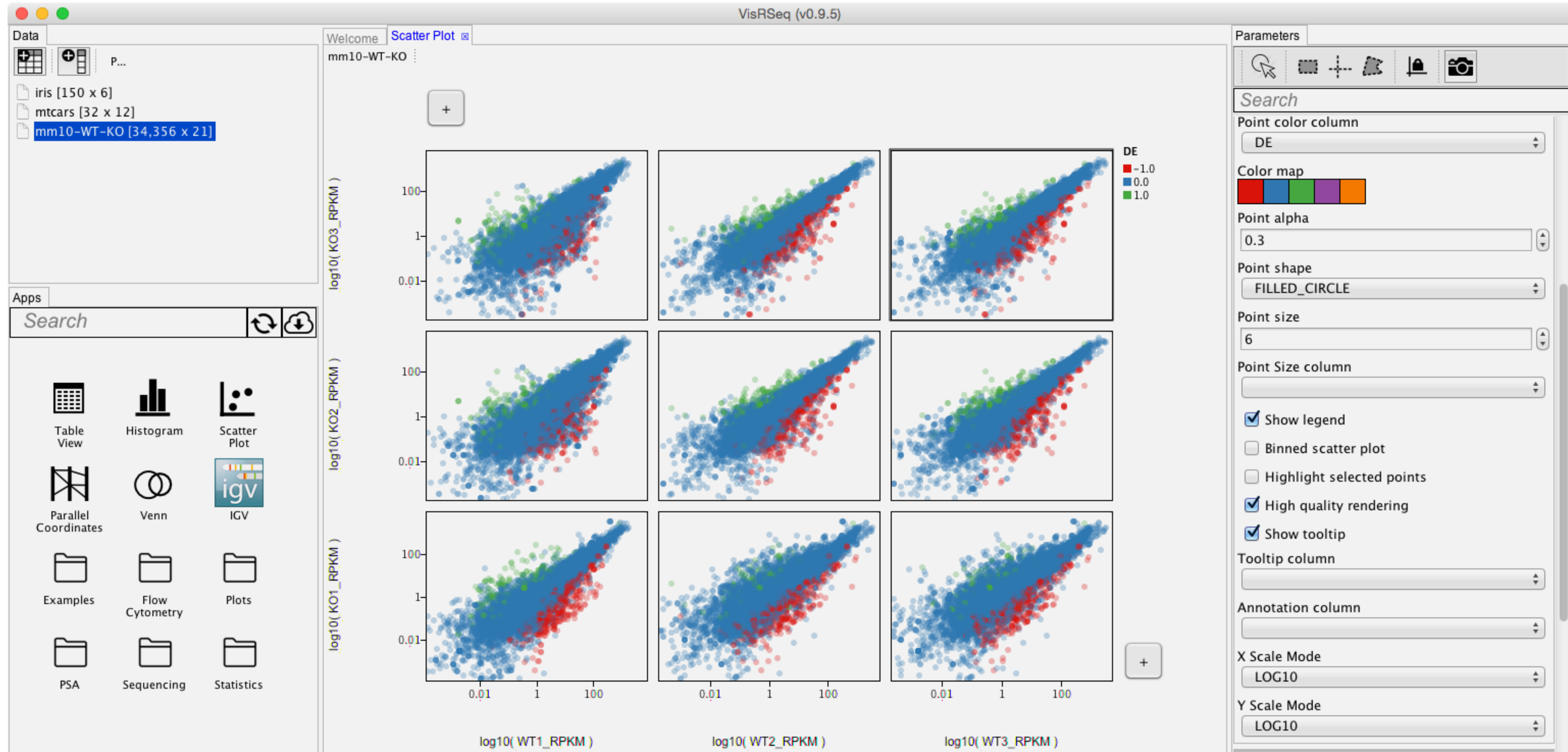


interface: parameters

parameters

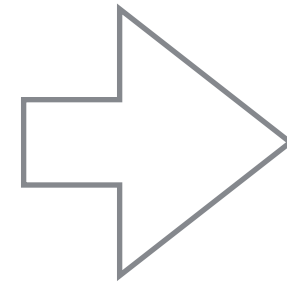
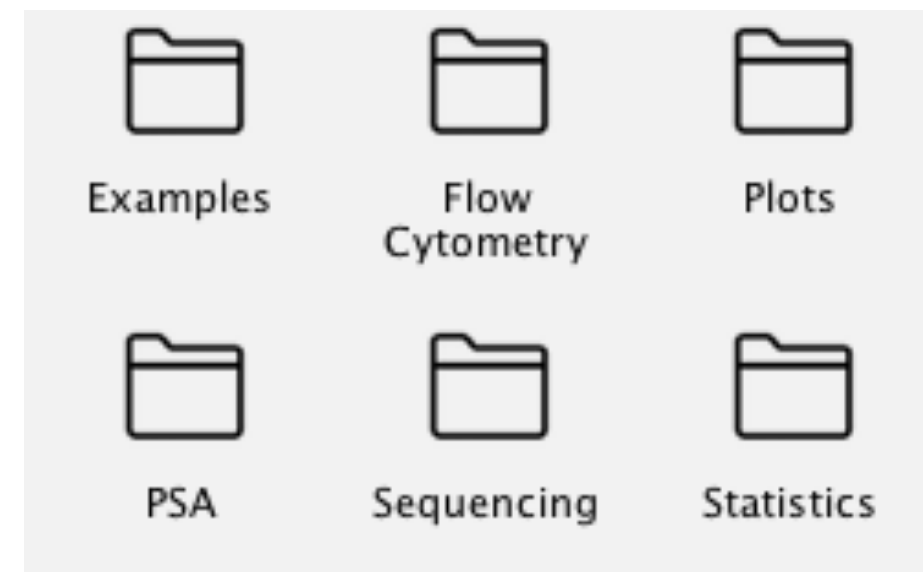


Apps: interactive

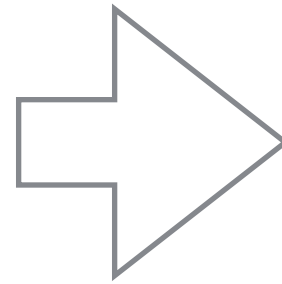
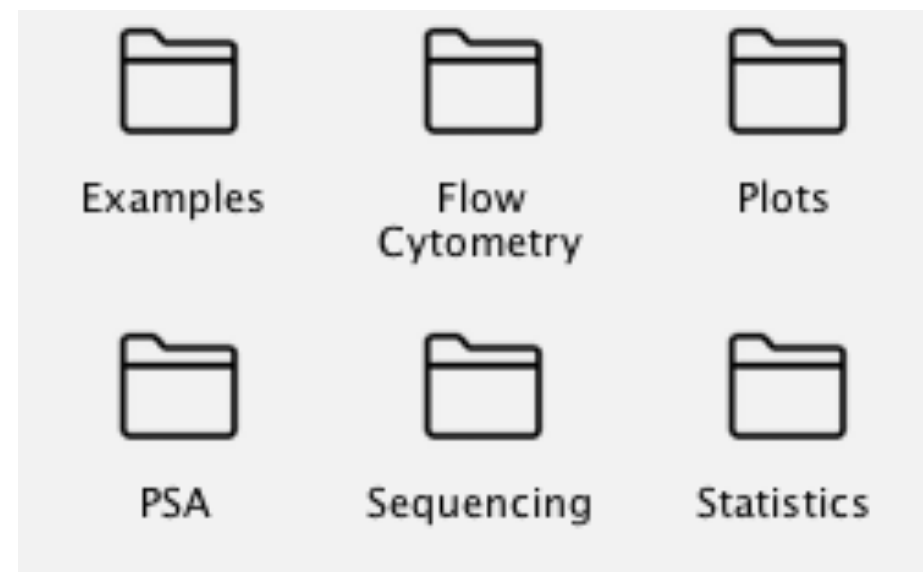


R - apps

R Apps



R Apps



[Plots]

BarPlot Boxplot Correlogram
DensityPlot DensityPlot2 DensityPlot3 heatmap
MosaicPlot OmicCircos PieChart Scatterplot 3D
Scatterplot ggplot ScreePlot ScreePlot2 Stacked Histogram

[Flow Cytometry]

flowClust flowMeans SamSPECTRAL summary

[Statistics]

Clustering Dimensional Analysis Distributions
Models Summary Tests

[RNA-seq]

baySeq Compute RLE DESeq
DESeq2 edgeR NOISeq sSeq
timeClip tweeDEseq VolcanoPlot

Anatomy of an R app

R code

kmeans.R

```
source("visrutils.R")  
visr.applyParameters()  
cluster_data<-subset(visr.input, select = param.columns)  
output.clusterid <- kmeans(cluster_data, param.k,  
                           algorithm = param.algorithm)$cluster  
plot(cluster_data, main = param.plot.title,  
      col = as.integer(output.clusterid))
```


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output.clusterid <- kmeans(cluster_data, param.k,  
                           algorithm = param.algorithm)$cluster  
plot(cluster_data, main = param.plot.title,  
      col = as.integer(output.clusterid))
```

parameters

kmeans.json

```
{ "label":"kmeans", "categories":[]  
  { "variables":{  
    "param.columns":  
    "param.k":  
    "param.algorithm":  
  
    "param.plot.title":  
    "output.clusterid":  
  }  
}
```

Anatomy of an R app

R code

kmeans.R

```
source("visrutils.R")  
visr.applyParameters()  
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      col = as.integer(output.clusterid))
```

parameters

kmeans.json

```
{ "label":"kmeans", "categories":[  
  { "variables":{  
    "param.columns":  
    "param.k":  
    "param.algorithm":  
  
    "param.plot.title":  
    "output.clusterid":  
  }  
}]
```

Anatomy of an R app

R code

kmeans.R

```
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visr.applyParameters()  
  
cluster_data<-subset(visr.input, select = param.columns)  
  
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                           algorithm = param.algorithm)$cluster  
plot(cluster_data, main = param.plot.title,  
      col = as.integer(output.clusterid))
```

parameters

kmeans.json

```
{ "label":"kmeans", "categories":[  
  { "variables":{  
    "param.columns": {"type":"multi-column-numerical"},  
    "param.k": {"type":"int", "default":3 },  
    "param.algorithm": {"items":["Hartigan-Wong", "Lloyd",  
                                "Forgy", "MacQueen"]},  
    "param.plot.title":{"default":"kmeans result"},  
    "output.clusterid":{"type":"output-column"}  
  }  
}]
```

Anatomy of an R app

R code

kmeans.R

```
source("visrutils.R")  
  
visr.applyParameters()  
  
cluster_data<-subset(visr.input, select = param.columns)  
  
output.clusterid <- kmeans(cluster_data, param.k,  
                           algorithm = param.algorithm)$cluster  
plot(cluster_data, main = param.plot.title,  
      col = as.integer(output.clusterid))
```

parameters

kmeans.json

```
{ "label":"kmeans", "categories":[  
  { "variables":{  
    "param.columns": {"type":"multi-column-numerical" },  
    "param.k": {"type":"int", "default":3 },  
    "param.algorithm": {"items":["Hartigan-Wong", "Lloyd",  
                                "Forgy", "MacQueen"]},  
    "param.plot.title":{"default":"kmeans result"},  
    "output.clusterid":{"type":"output-column"}  
  }  
}]
```

Anatomy of an R app

R code

kmeans.R

```
source("visrutils.R")  
visr.applyParameters()  
cluster_data<-subset(visr.input, select = param.columns)  
output.clusterid <- kmeans(cluster_data, param.k,  
                           algorithm = param.algorithm)$cluster  
plot(cluster_data, main = param.plot.title,  
      col = as.integer(output.clusterid))
```

parameters

kmeans.json

```
{ "label":"kmeans", "categories":[  
  { "variables":{  
    "param.columns": {"type":"multi-column-numerical" },  
    "param.k":       {"type":"int", "default":3 },  
    "param.algorithm": {"items":["Hartigan-Wong", "Lloyd",  
                                "Forgy", "MacQueen"]},  
    "param.plot.title":{"default":"kmeans result"},  
    "output.clusterid":{"type":"output-column"}  
  }  
}]
```

auto generated UI

kmeans

columns

- index
- Sepal.Length
- Sepal.Width
- Petal.Length
- Petal.Width

k

3

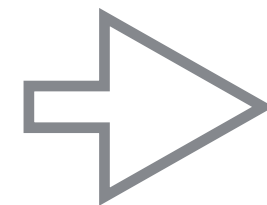
algorithm

Hartigan-Wong

plot title

kmeans result

clusterid



Anatomy of an R app

R code

kmeans.R

```
source("visrutils.R")  
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      col = as.integer(output.clusterid))
```

parameters

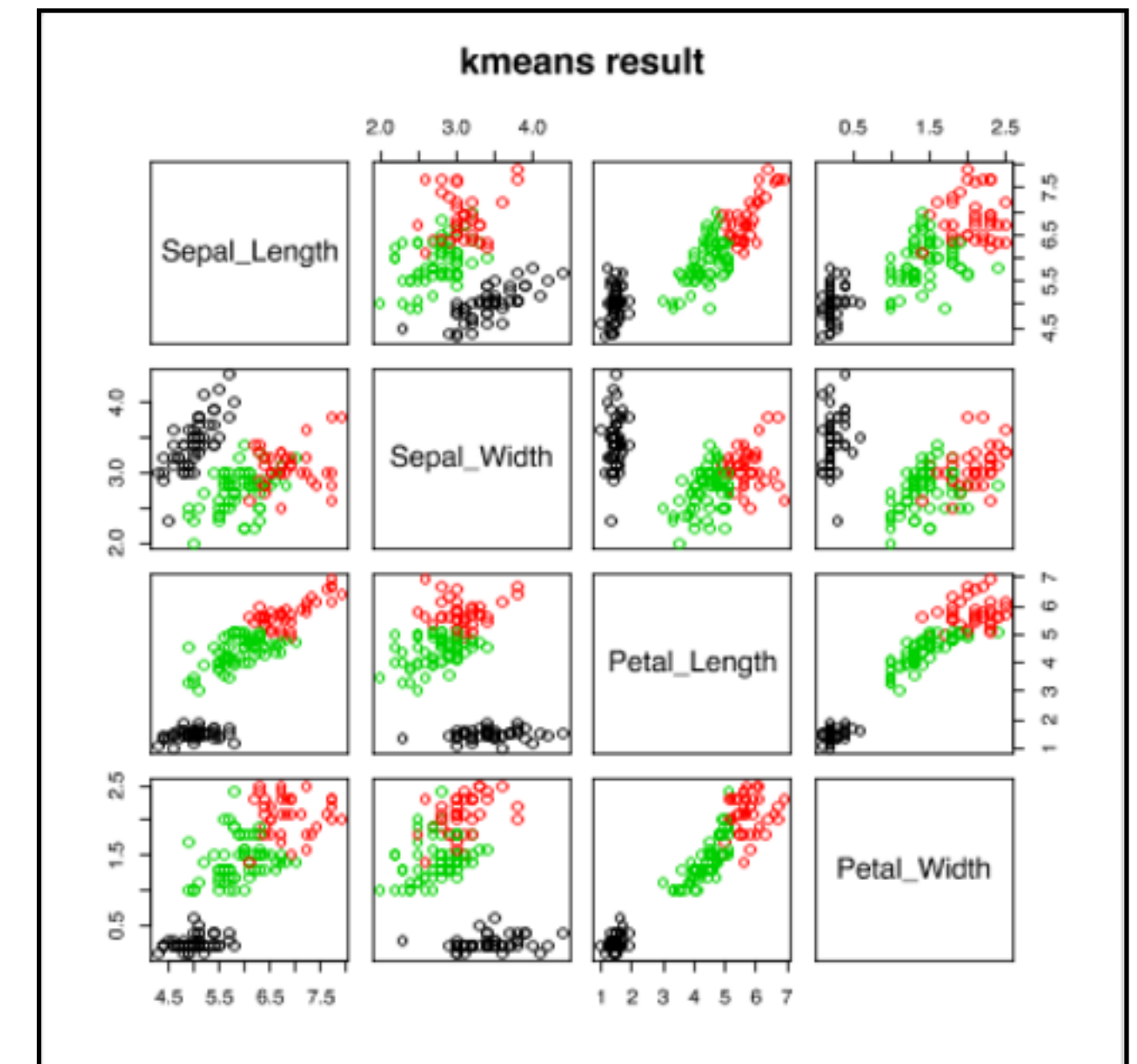
kmeans.json

```
{ "label":"kmeans", "categories":[  
  { "variables":{  
    "param.columns": {"type":"multi-column-numerical" },  
    "param.k": {"type":"int", "default":3 },  
    "param.algorithm": {"items":["Hartigan-Wong", "Lloyd",  
                                "Forgy", "MacQueen"]},  
    "param.plot.title":{"default":"kmeans result"},  
    "output.clusterid":{"type":"output-column"}  
  }  
}]
```

auto generated UI

The screenshot shows a graphical user interface for a kmeans application. The window title is "kmeans". It contains several interactive elements: a list of columns with "index" selected, a numeric input for "k" set to 3, a dropdown menu for "algorithm" set to "Hartigan-Wong", a text input for "plot title" containing "kmeans result", and a text input for "clusterid".

output



creating R apps: variable types

variable type
int
double
boolean
string
string with items
filename
color
range-int
range-double
column
column-numerical
multi-column
multi-column-numerical
ouput-column
output-table

20

show percentage

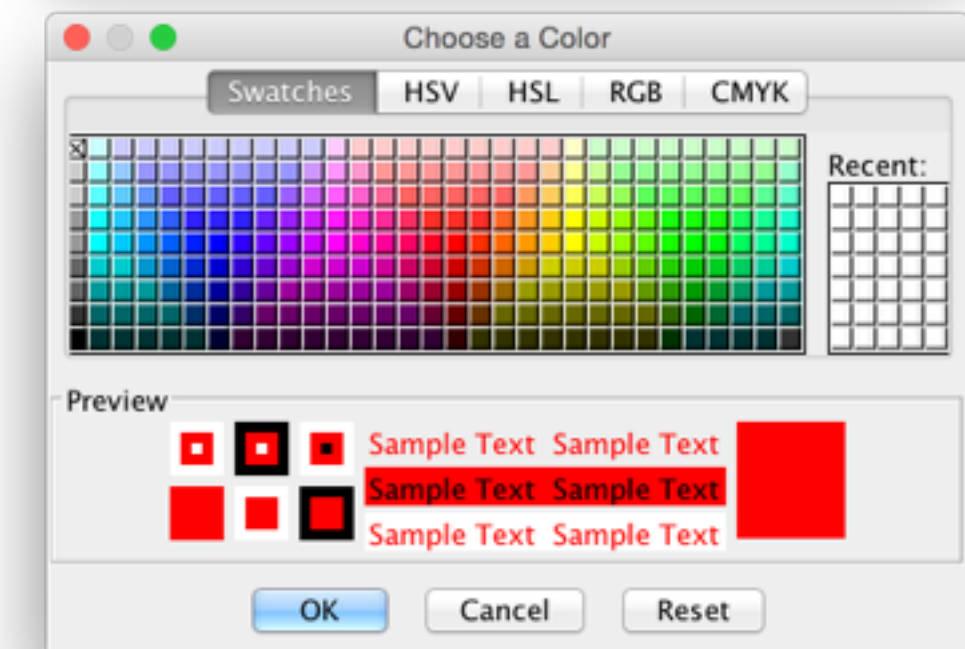
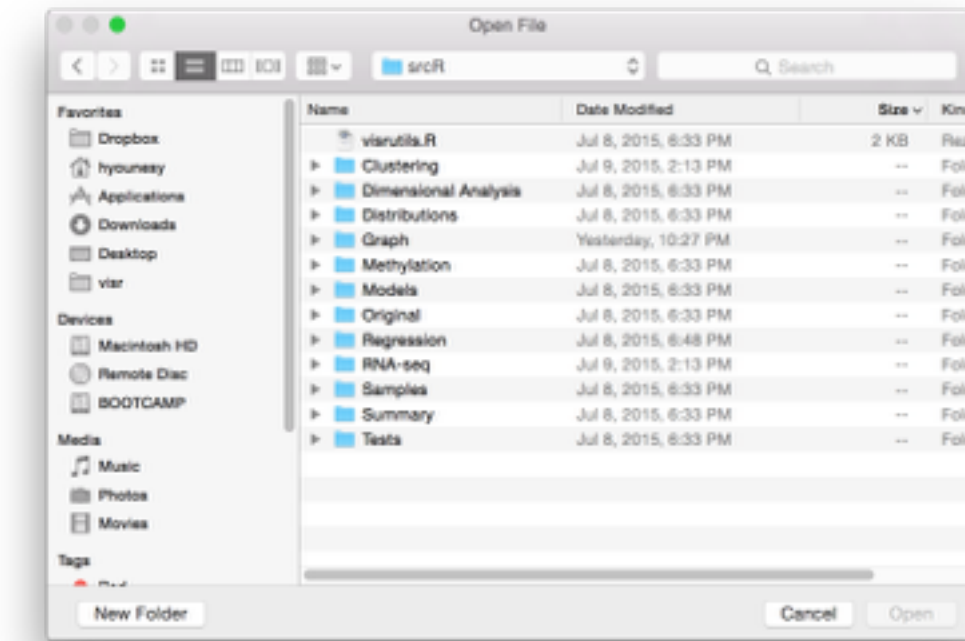
Plot Title

Hartigan-Wong

...

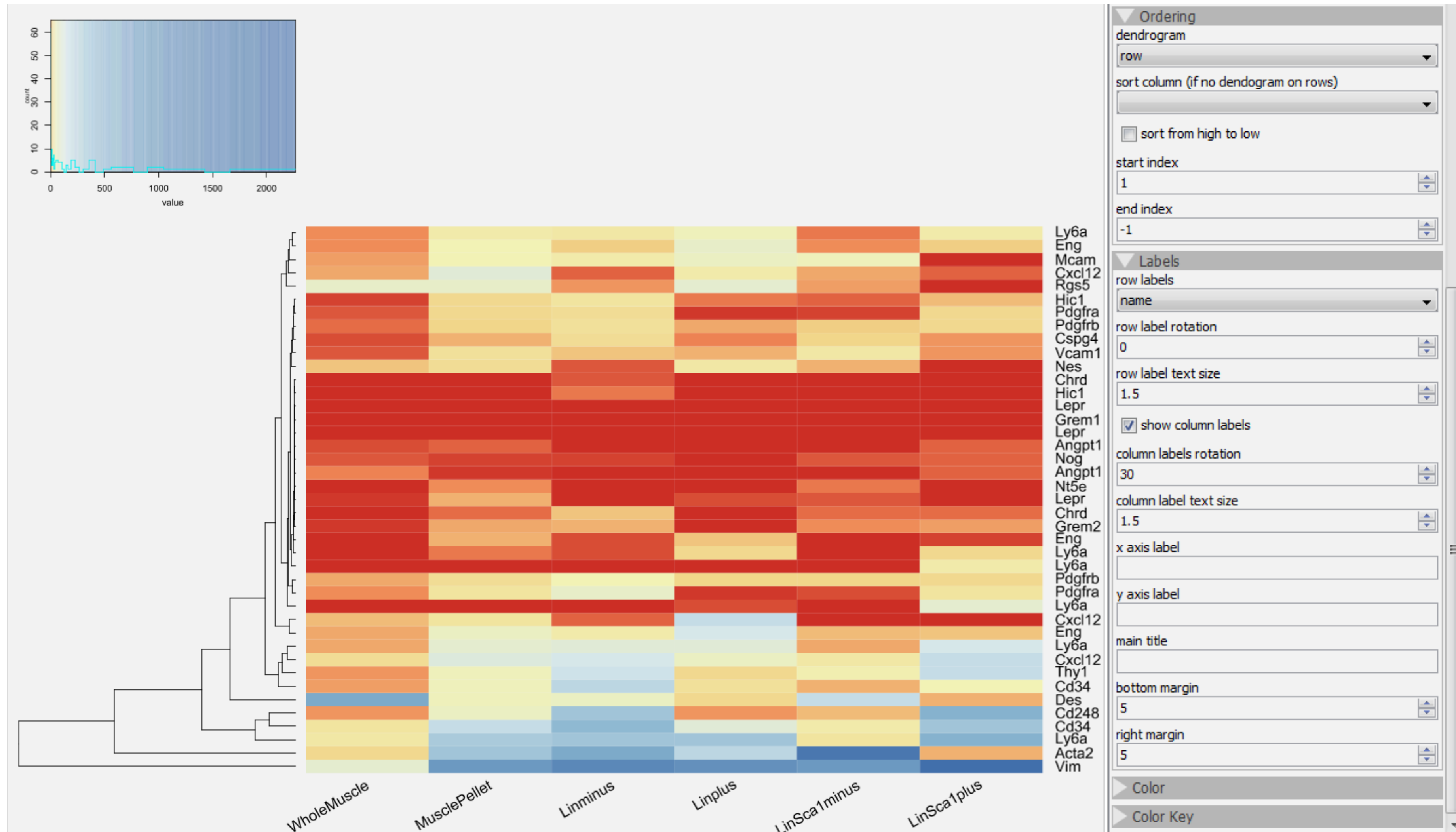
RNA-seq_CASTEij
RNA-seq_C57BL6J

chr
✓ chr
name
RNA-seq_CASTEij
RNA-seq_C57BL6J

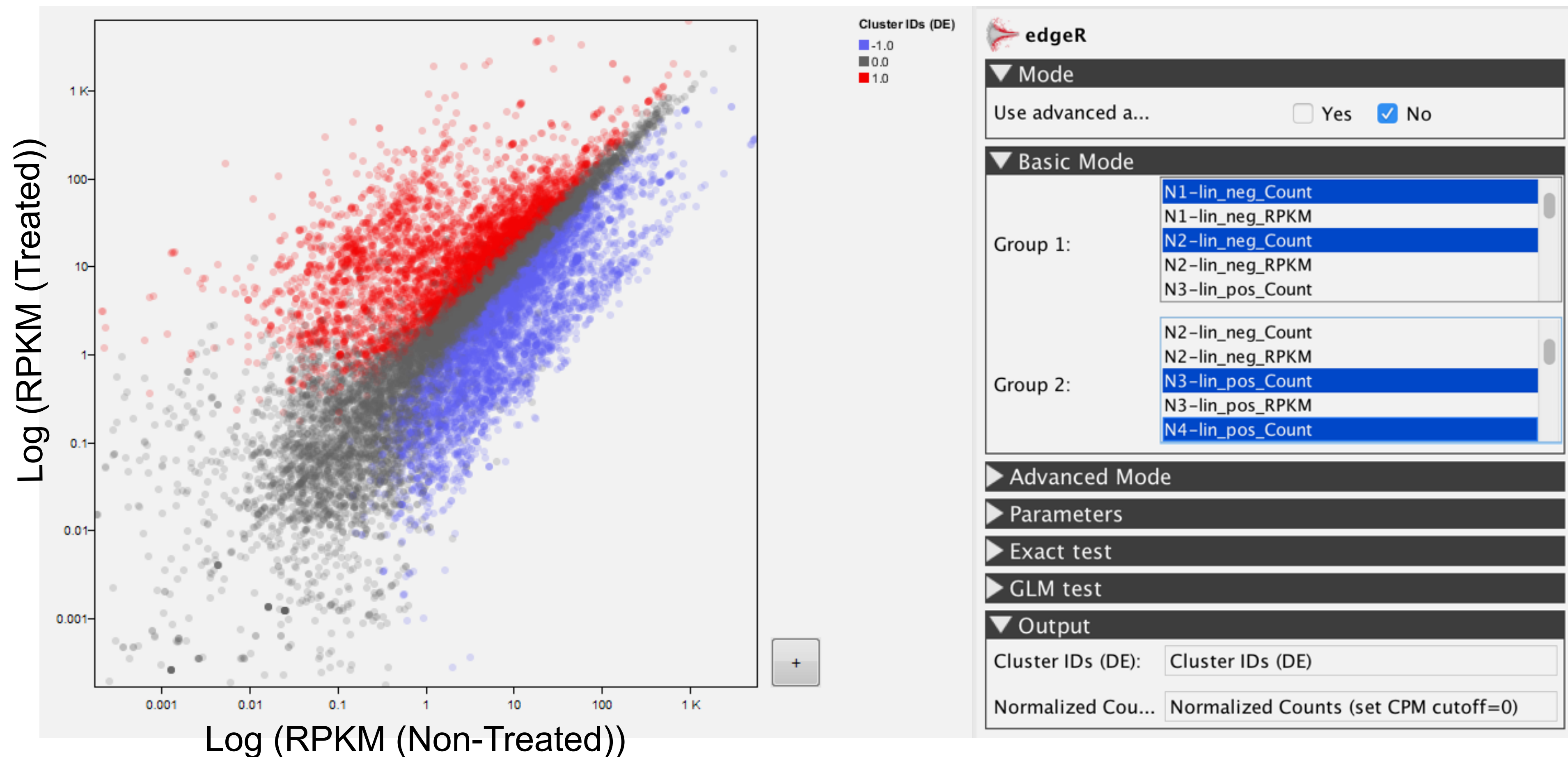


`visr.message(message, type)`

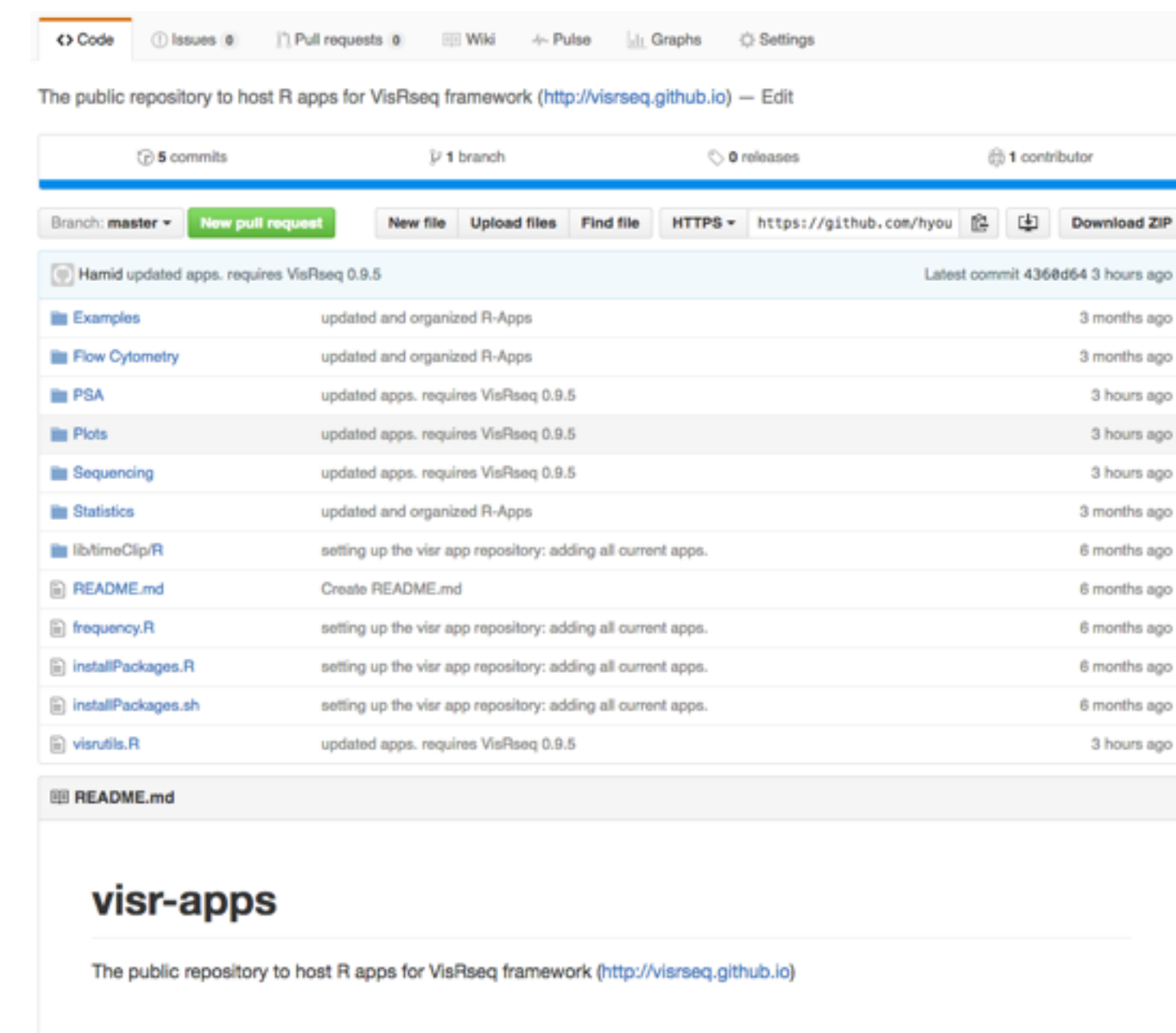
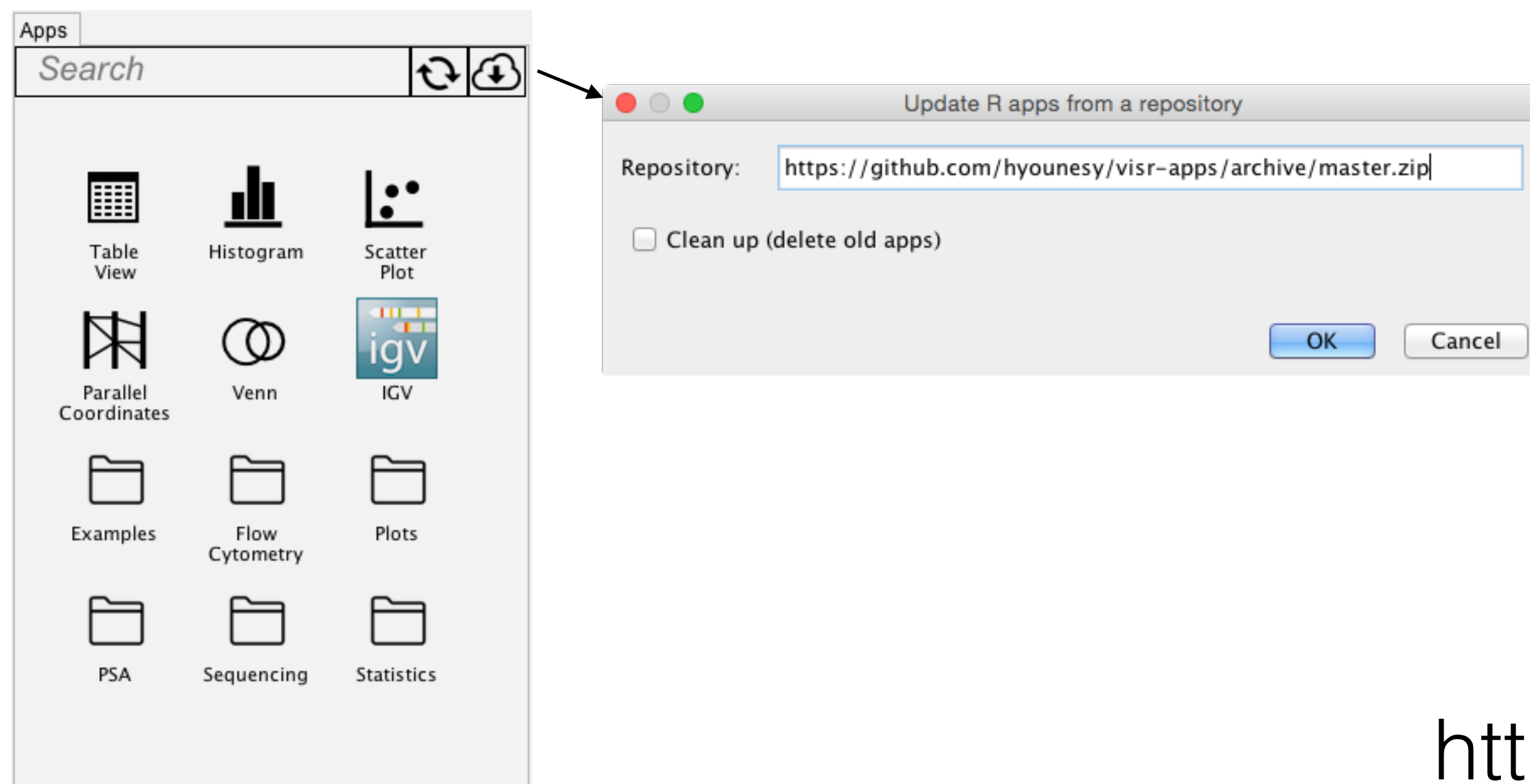
R – Apps example: heatmap



R – Apps example: edgeR



creating R apps: sharing

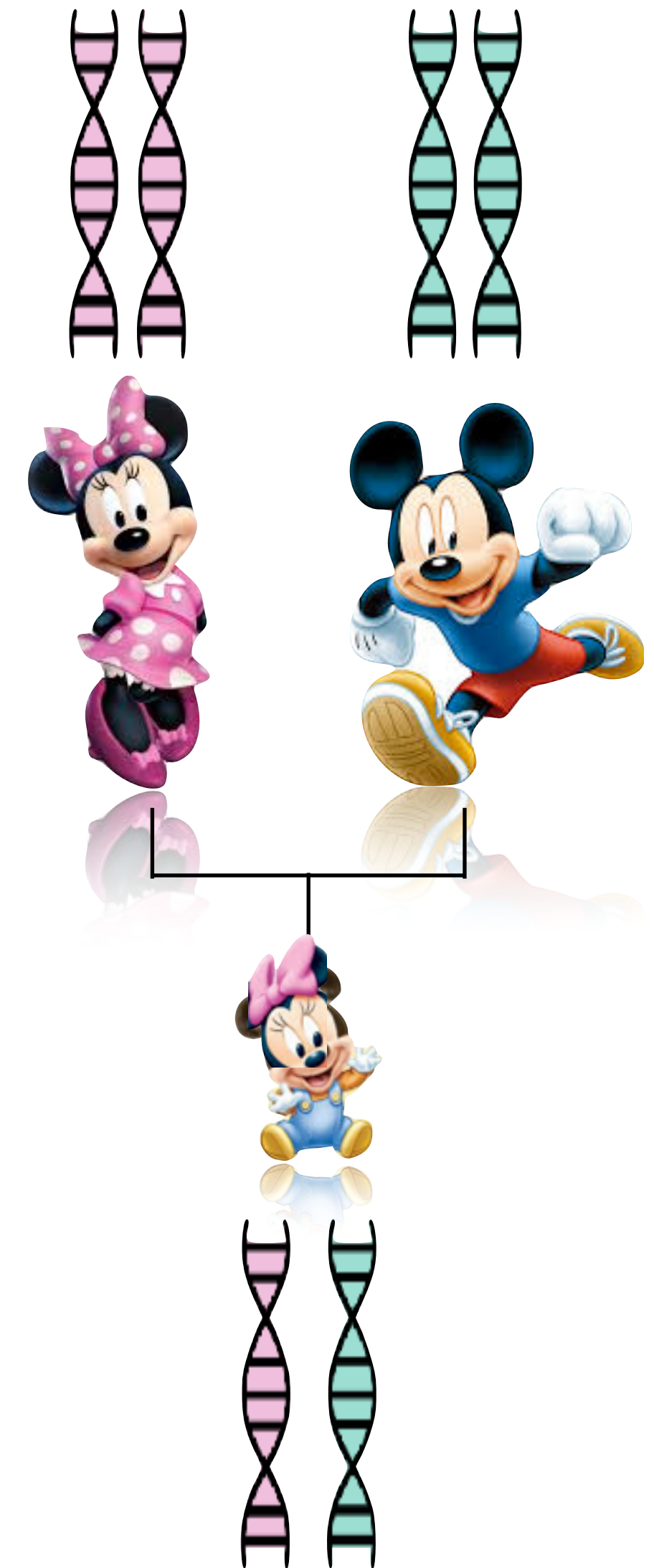


<https://github.com/hyounesy/visr-apps/>

case study

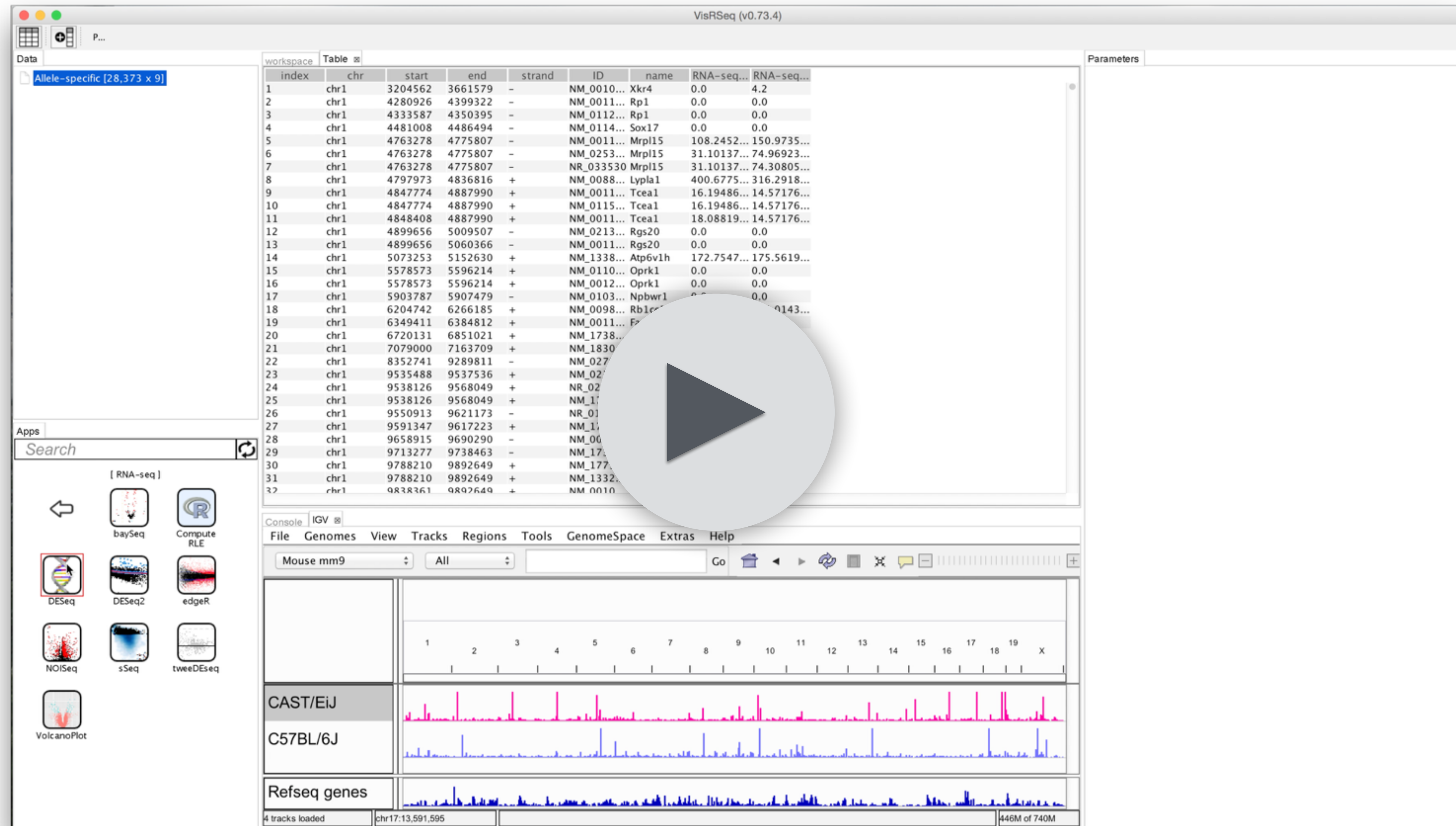
case study

- allele specific gene expression (RNA-seq)
- F1 hybrid mice: CAST + C57
- allelic RNA-seq created using ALEA^[1]



[1] H. Younesy, et.al.: ALEA: a toolbox for allele-specific epigenomics analysis. *Bioinformatics* 30(8), 1172–1174 (2014)

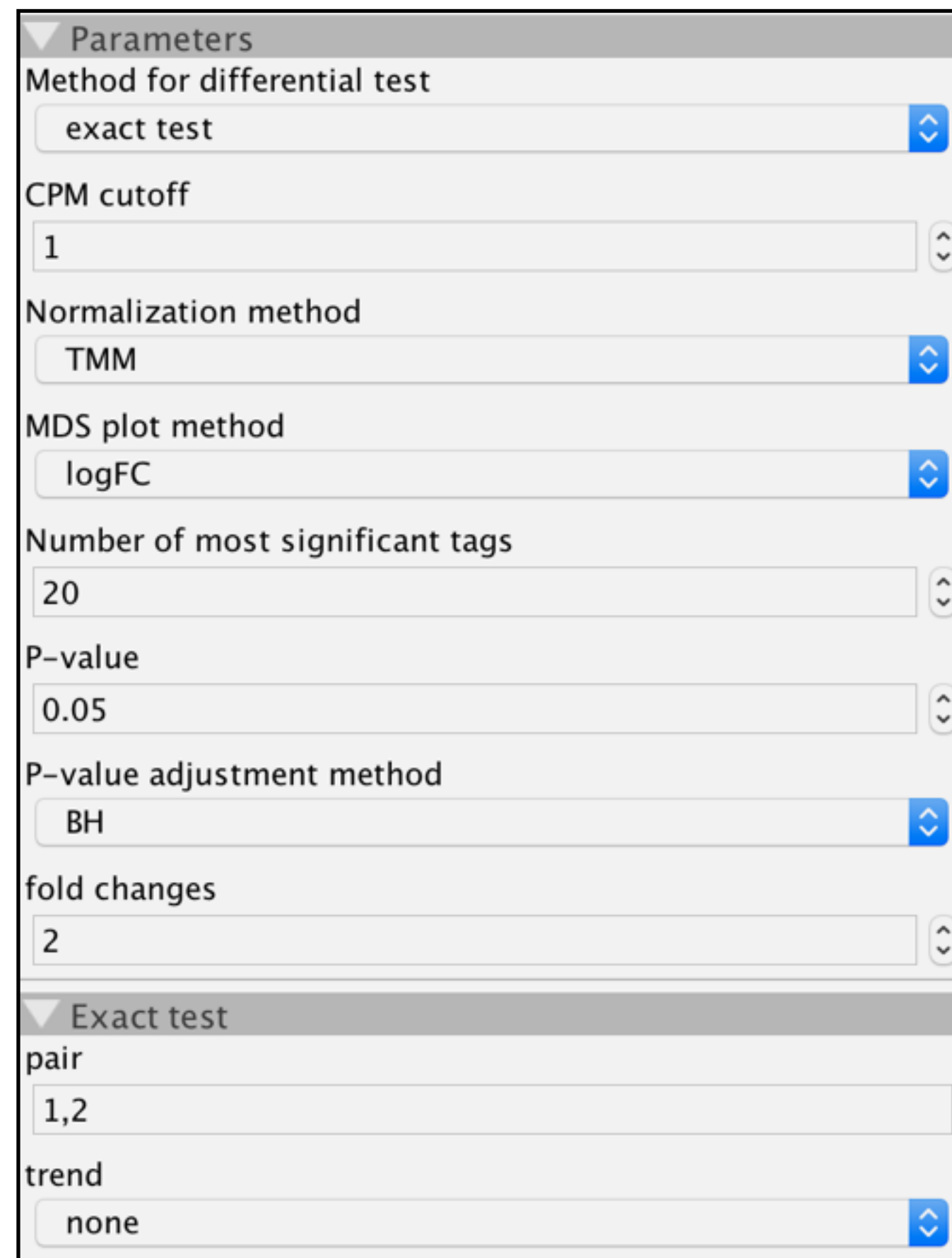
case study: allele specific expression



parameter space
exploration

parameter space exploration: motivation

what should be the parameter values?



The image shows a screenshot of a software interface with a 'Parameters' section. The parameters are as follows:

- Method for differential test: exact test
- CPM cutoff: 1
- Normalization method: TMM
- MDS plot method: logFC
- Number of most significant tags: 20
- P-value: 0.05
- P-value adjustment method: BH
- fold changes: 2

Below the 'Parameters' section is an 'Exact test' section with the following settings:

- pair: 1,2
- trend: none

parameter space exploration: interface

Parameter exploration

Parameters

Method for differential test
exact test

CPM cutoff
1

Normalization method
TMM

MDS plot method
logFC

Number of most significant tags
20

P-value
0.05

P-value adjustment method
BH

fold changes
2

Exact test

pair
1,2

trend
none

parameter space exploration: interface

Parameter exploration

Parameters

Method for differential test
exact test

CPM cutoff
1

Normalization method
TMM

MDS plot method
logFC

Number of most significant tags
20

P-value
0.05

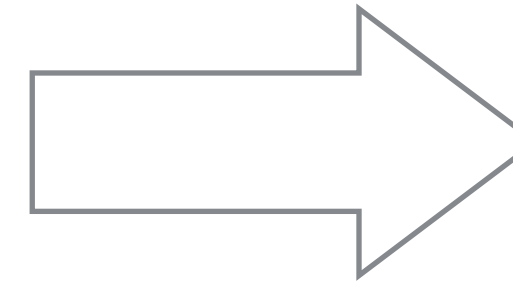
P-value adjustment method
BH

fold changes
2

Exact test

pair
1,2

trend
none



Parameters

Method for differential test
exact test

CPM cutoff: 0.509

Normalization ...
TMM RLE upperquart none

MDS plot metho...
logFC bcv

Number of most... 20

P-value: 0.001

P-value adjustm...
none BH fdr BY holm

fold changes: 1.572

Exact test

pair: 1,2

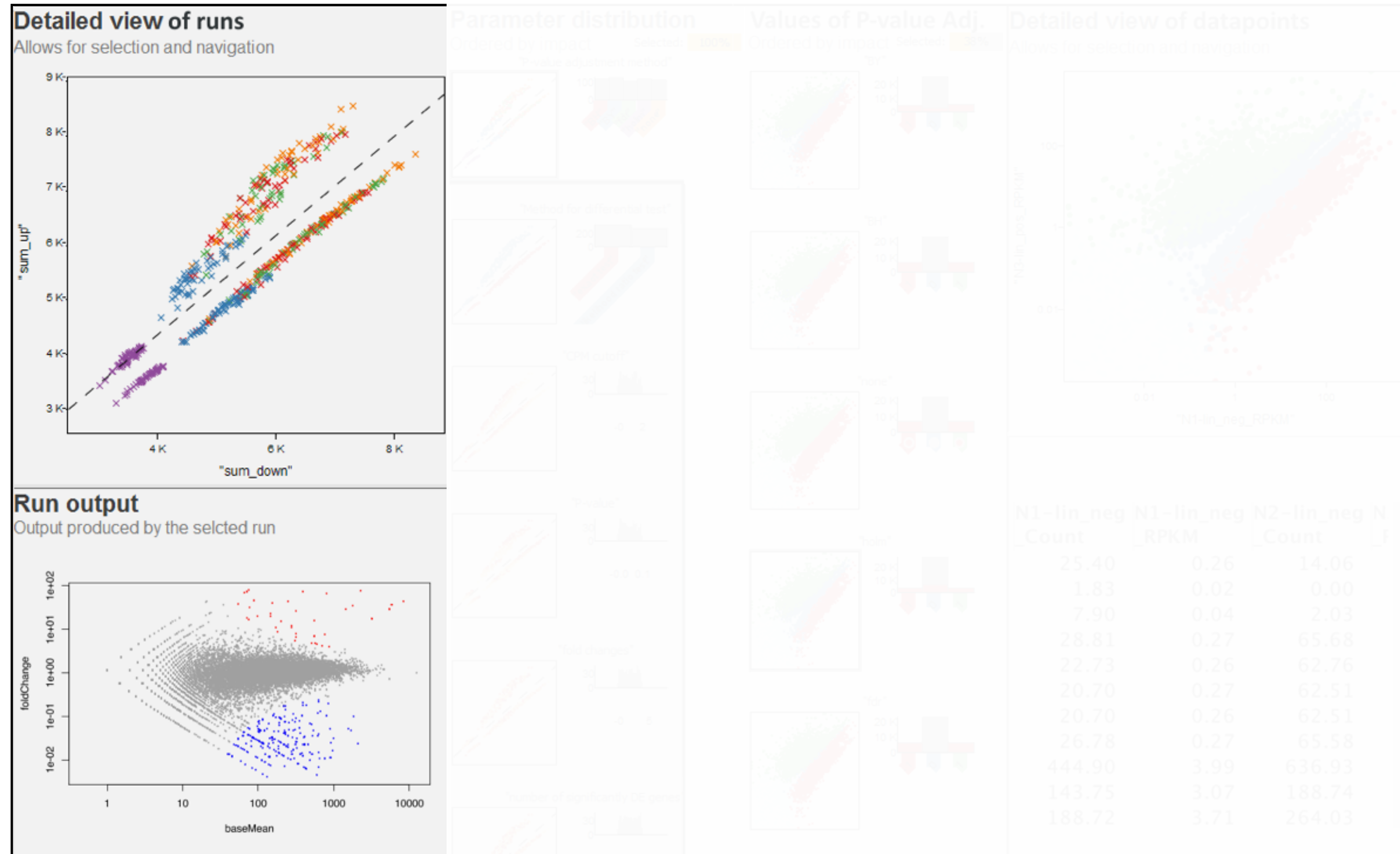
trend:
none movingave loess

GLM test

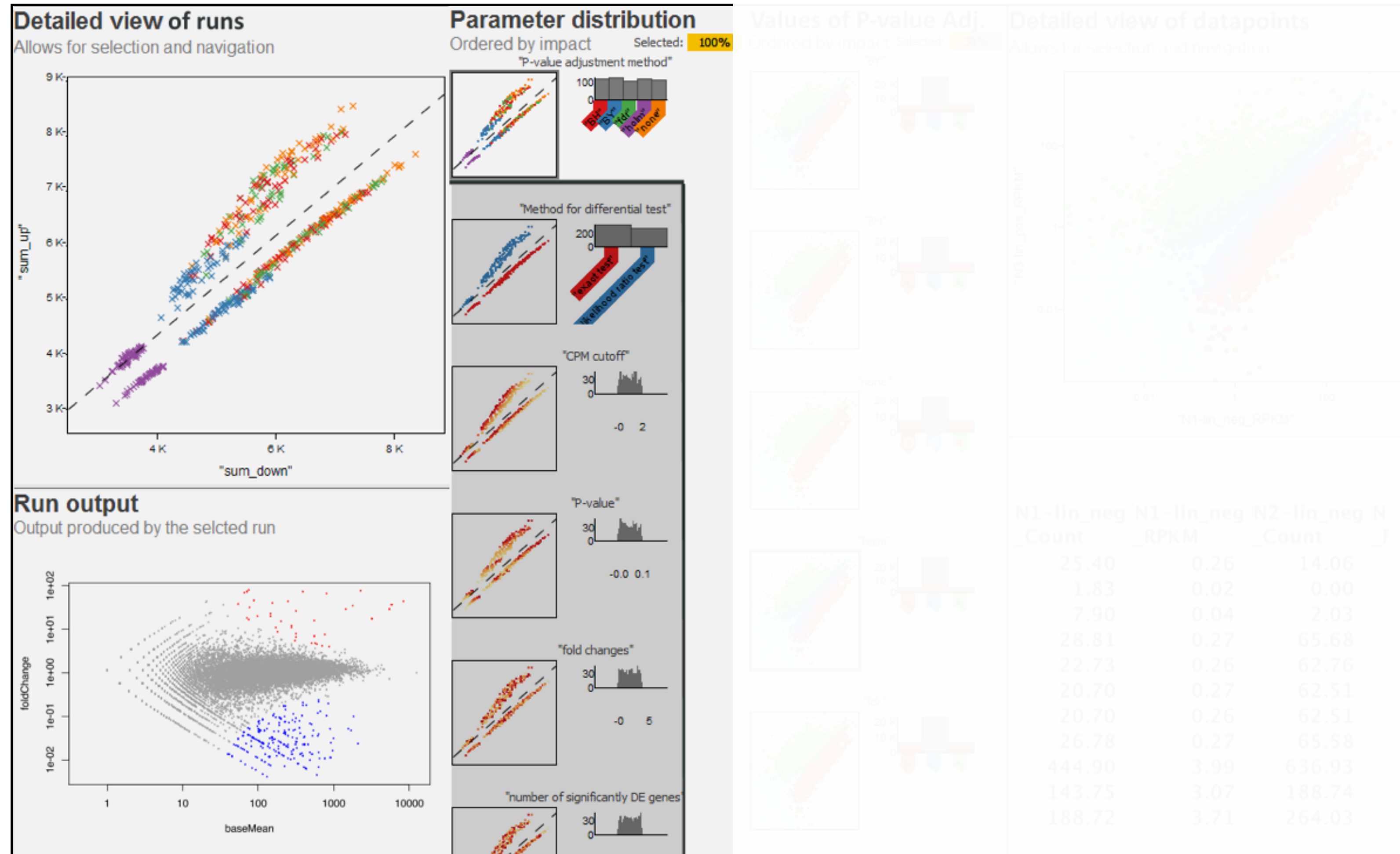
Runs Runs: 1 Run

✓ Runs Until

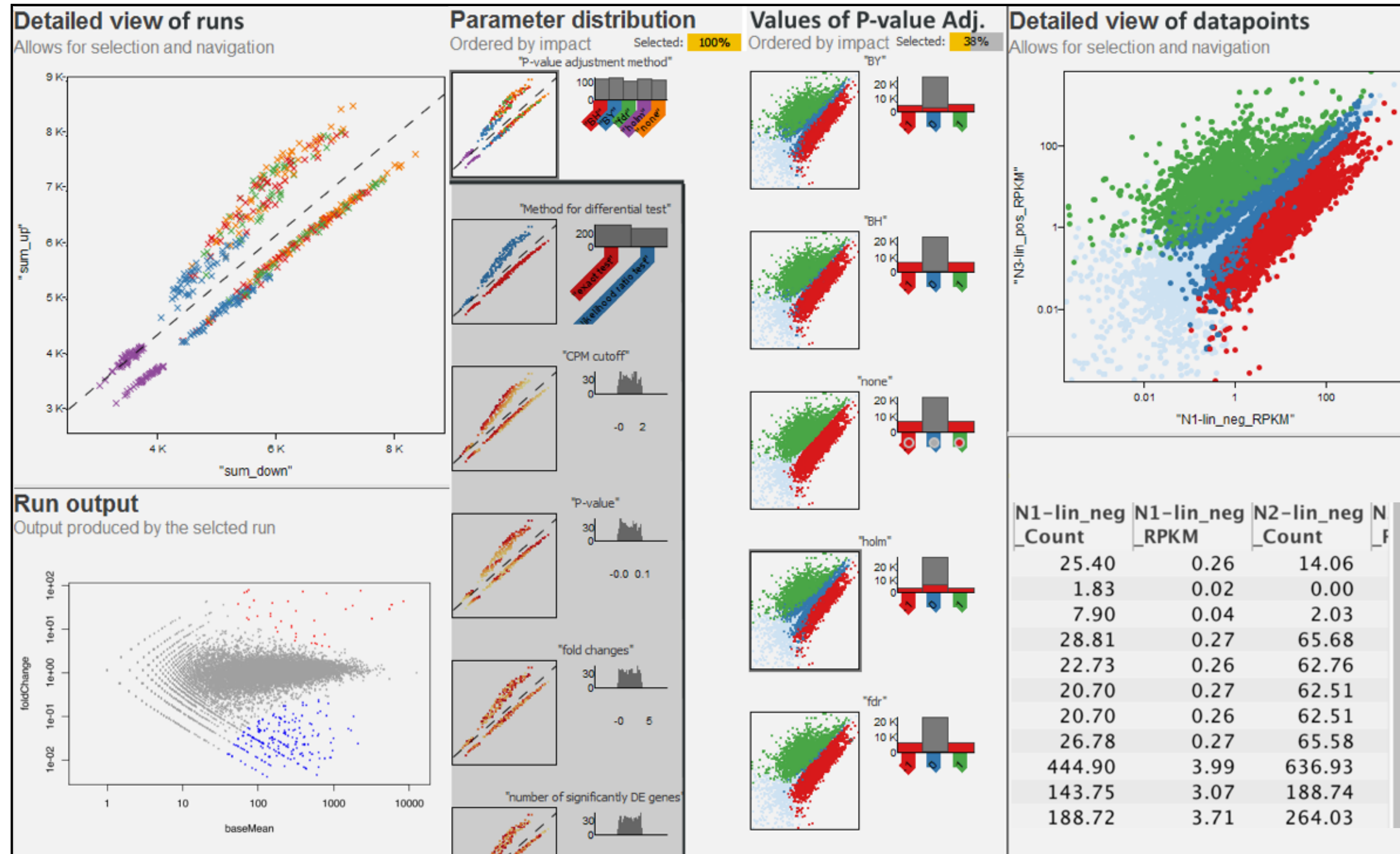
parameter space exploration: result



parameter space exploration: result



parameter space exploration: result



conclusions

limitations

- Scalability: few million rows and 30-40 columns of sequencing data
- cannot run R-apps in parallel
- desktop only

future work

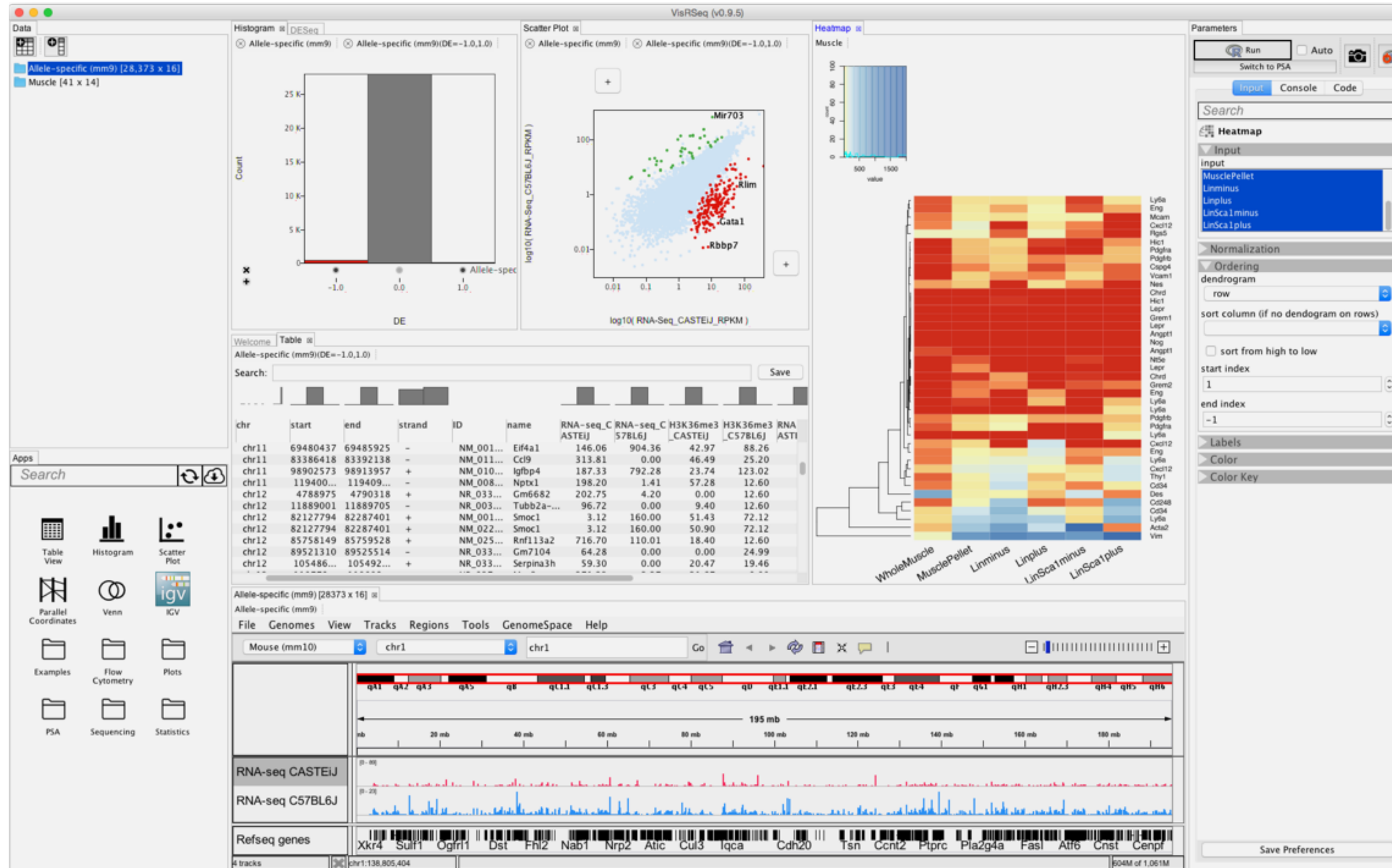
- general purpose framework: Apps for other domains (e.g. Proteomics, etc.)
- workflow designer : link several apps to create “macro” apps.
- reproducibility: provenance / parameter exploration

acknowledgement

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- BC Genome Sciences Centre
- Lorincz lab, Underhill lab, Rossi lab, Mager lab



questions?



visrseq.github.io
(please email for latest version)