

# **ExpVar: Gene Expression and Genetic Variants**

**Data Analysis And Visualization R package**

**EXPviz, SNPviz and CNVviz apps**

# **EXPviz App**

**For genetic expression data  
visualization**

# 1. EXPviz

EXPviz

Import Data

Count Data

Expression

Ontology

Import count File

Import the count csv file here  
count.csv

Browse...

count.csv

Upload complete

Import metadat File

Import the metadata txt file here  
metadata.txt

Browse...

No file selected

Visualize csv file

Show 10 entries

Search:

	gene	SRR11426824	SRR11426825	SRR11426826	SRR11426821	SRR11426822	SRR11426823
1	1	3	5	5	24	15	29
2	10	0	0	0	0	1	0
3	100	315	286	346	104	122	101
4	1000	1055	1235	1109	344	304	263
5	10000	824	844	738	207	163	145
6	100008586	0	0	0	0	0	0
7	100009676	27	27	30	77	42	59
8	10001	443	446	430	38	41	27
9	10002	46	44	48	11	12	6
10	10003	34	36	26	161	140	148

Showing 1 to 10 of 23,459 entries

Previous

1

2

3

4

5

...

2,346

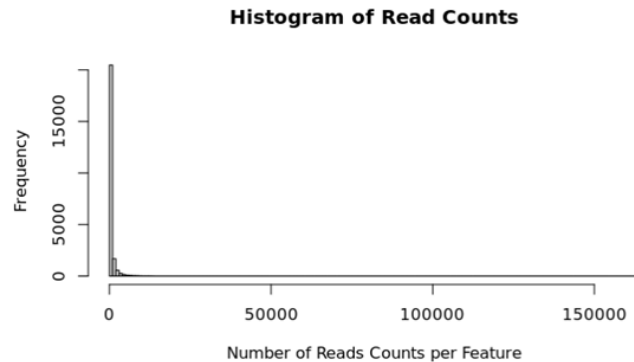
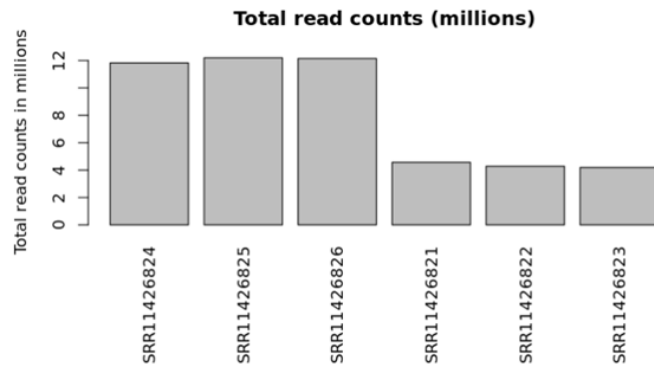
Next

Expression

Ontology

Read\_Count1

Before log2 transformation

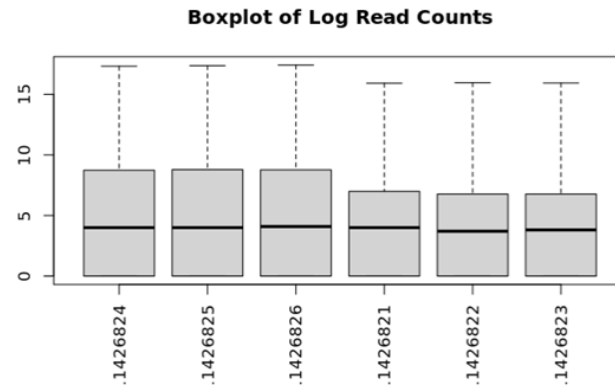
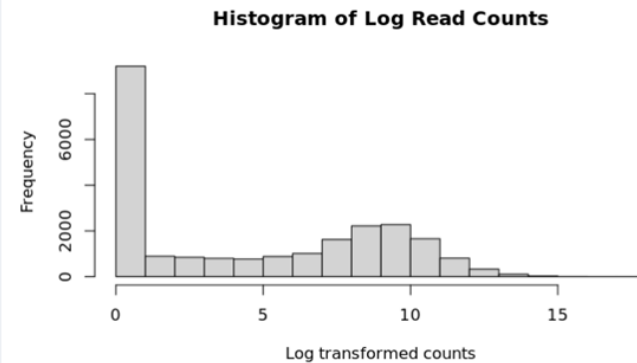


Read\_Count2

density plot

scatter plot

After log2 transformation

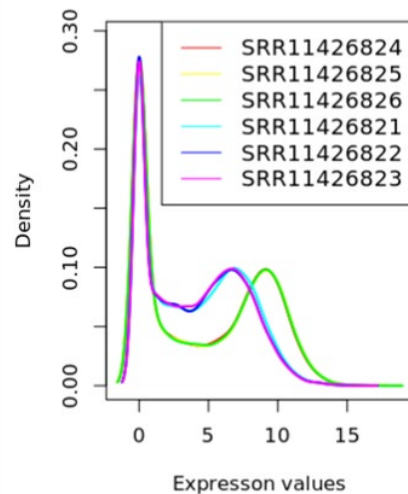


Read\_Count1

Read Count before log2 transformation

Before log2 transformation

### Distribution of transformed d



Read\_Count2

density plot

scatter plot

Read Count after log2 transformation

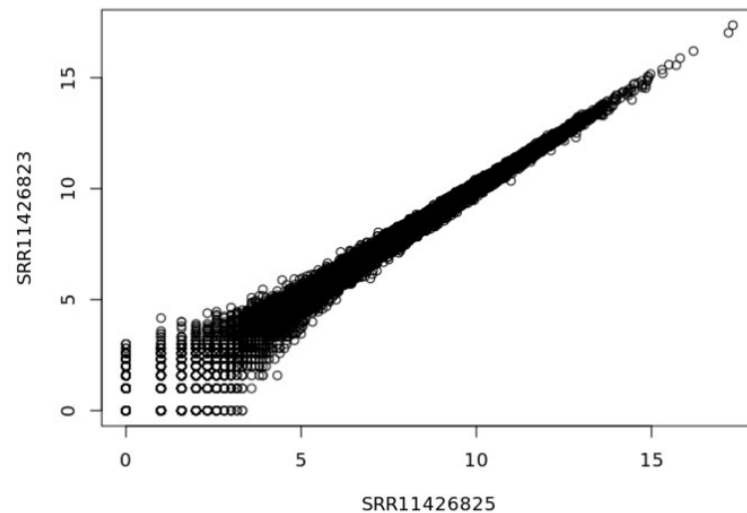
scatter plot

sample ID

eg: SRR11426825

sample ID

eg: SRR11426823



- ⌂ Import Data
- ⌂ Count Data
- ⌂ Expression
- ⌂ Ontology

expression

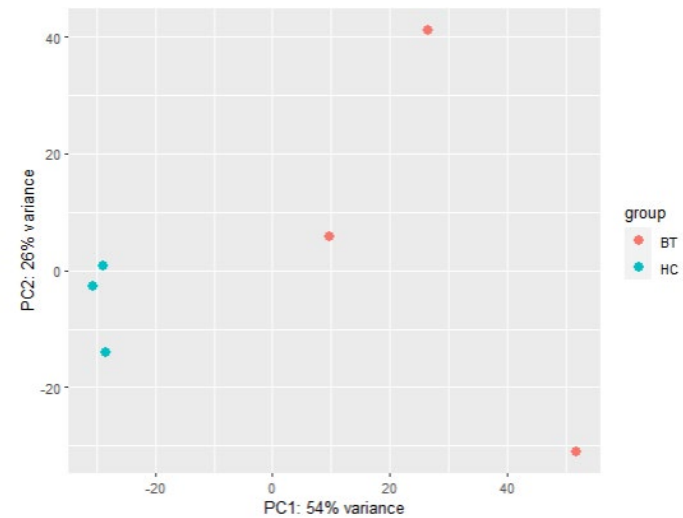
P value

0.05

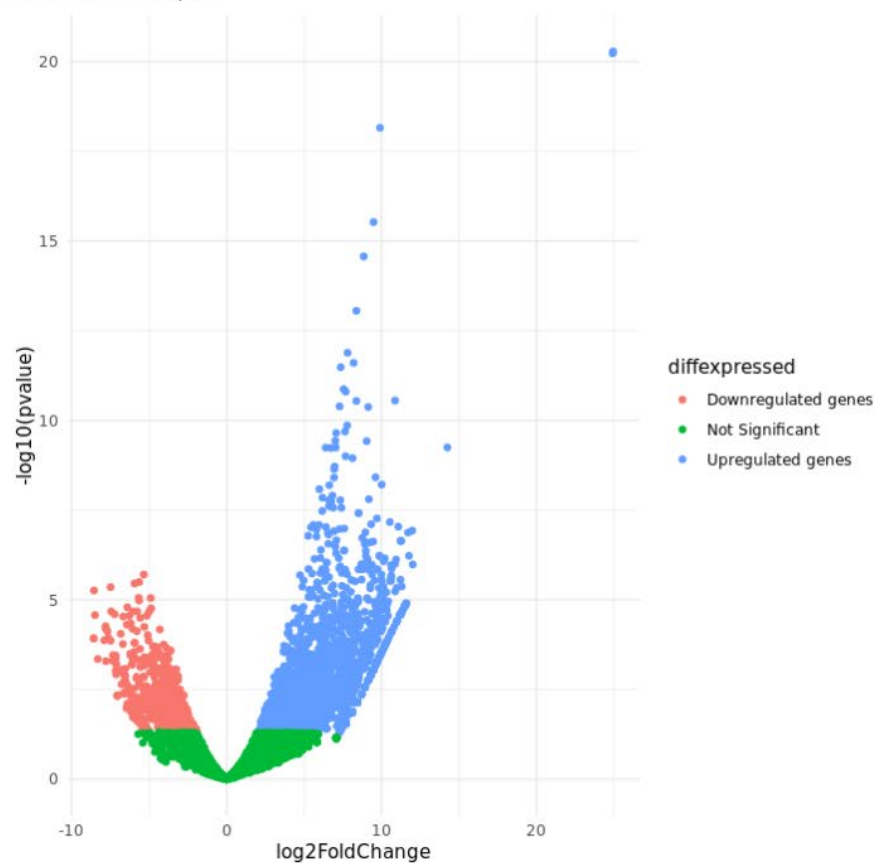
LogFC

2

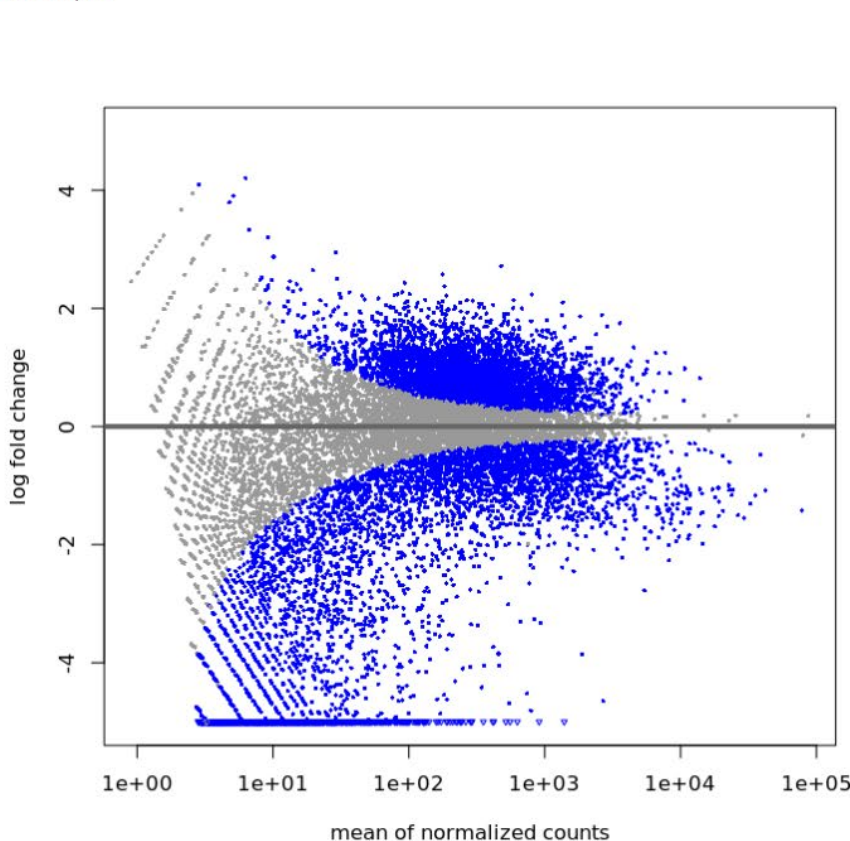
PCA PCA plot

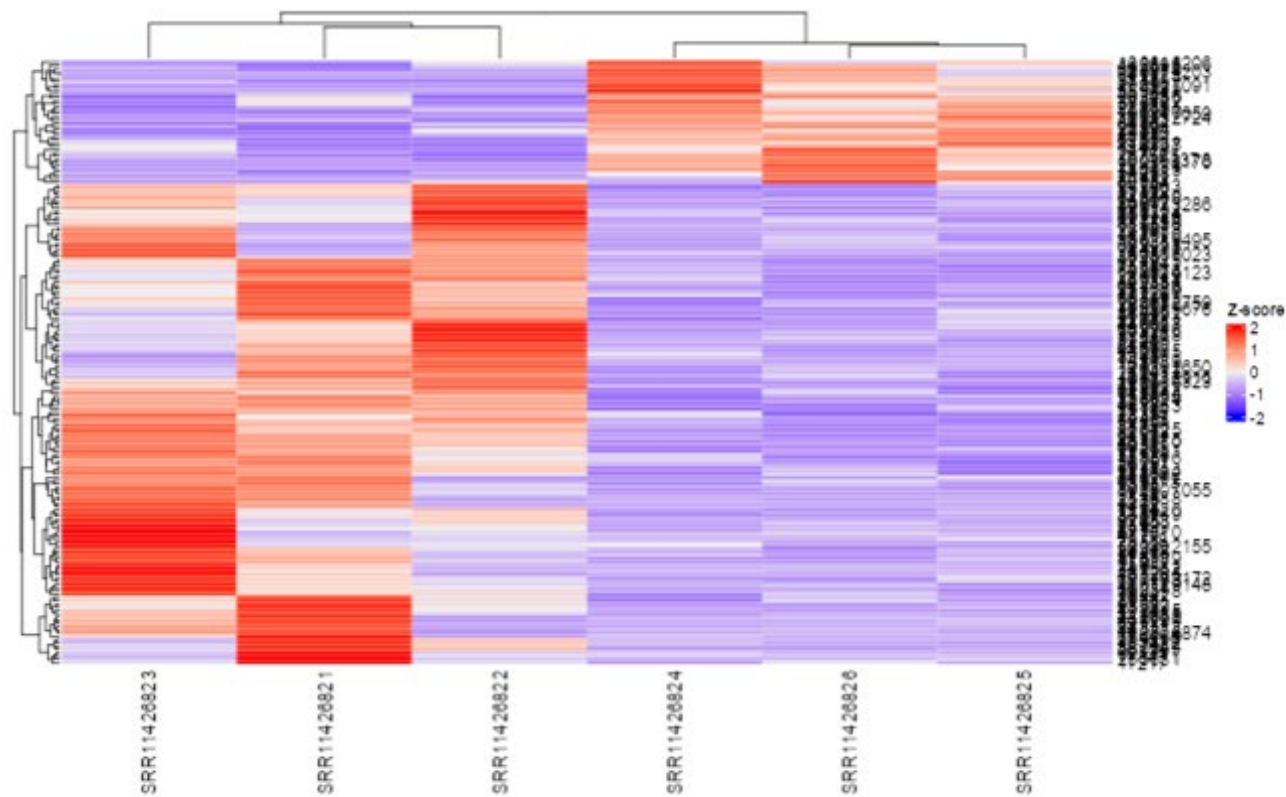


volcano Volcano plot



MA MA plot







- Import Data
- Count Data
- Expression
- Ontology

## Filters

Genes to analyze Note: DEGs= Differentially expressed genes

## Choose gene group

all DEGs

all DEGs

Upregulated genes

Downregulated genes

## Top Ontologies

20

## Biological Processes

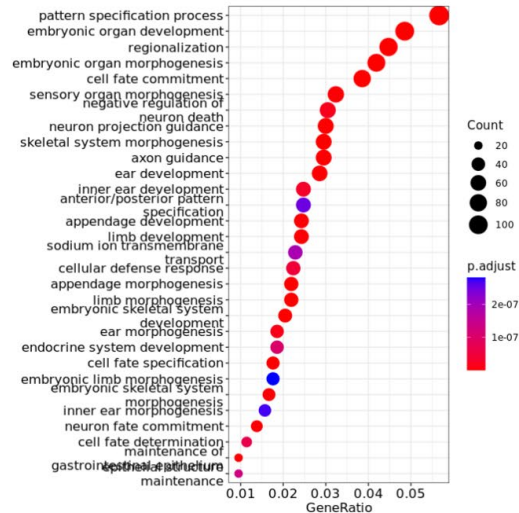
## Molecular Functions

## Cellular Component

Barplot1 BP Barplot



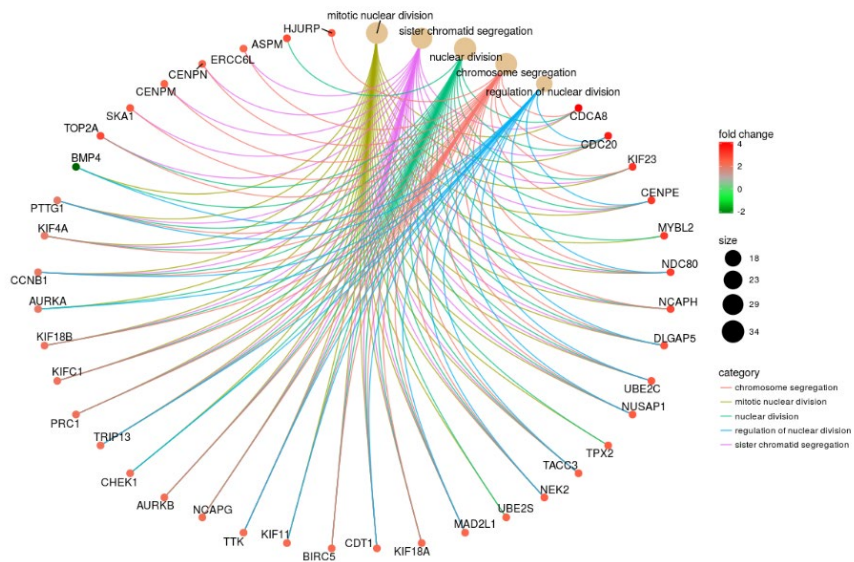
Dotplot1 BP dotplot



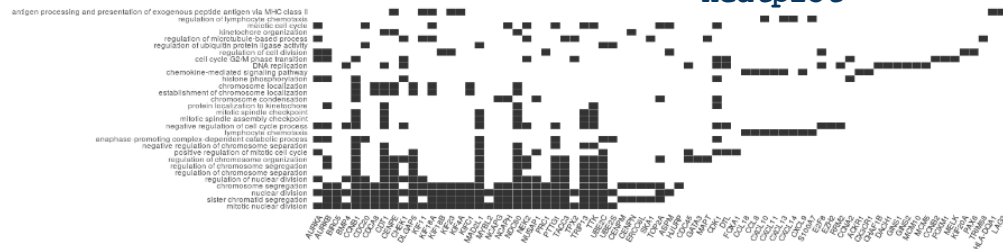
## Ontology

## Gene-Concept Network

## cnetplot

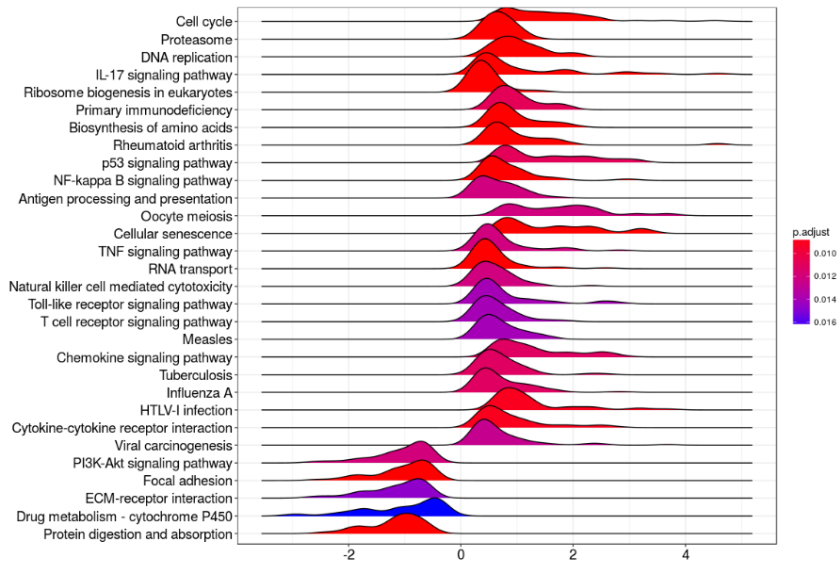


## heatplot

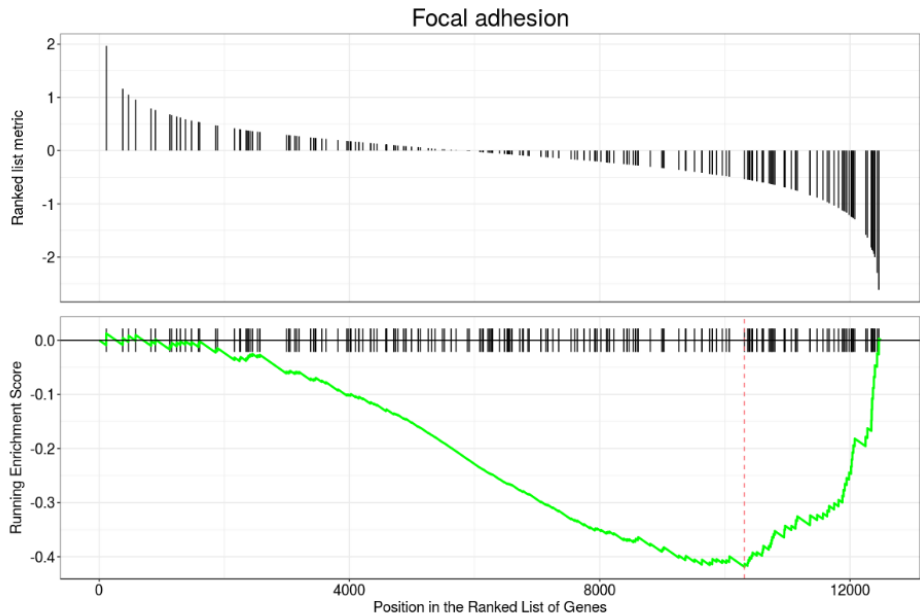


# expression distribution of GSEA result

ridgeplot



gseaplot



# **SNPviz App**

**For SNP data visualization**

## 2. SNPviz

SNPviz

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⌕ Data Import

⌕ SNP Data

⌕ Amino Acid Changes

### Visualize SNPs data

Import File

—

Import the variant data vcf file here

**Upload vcf file**

Browse...

No file selected

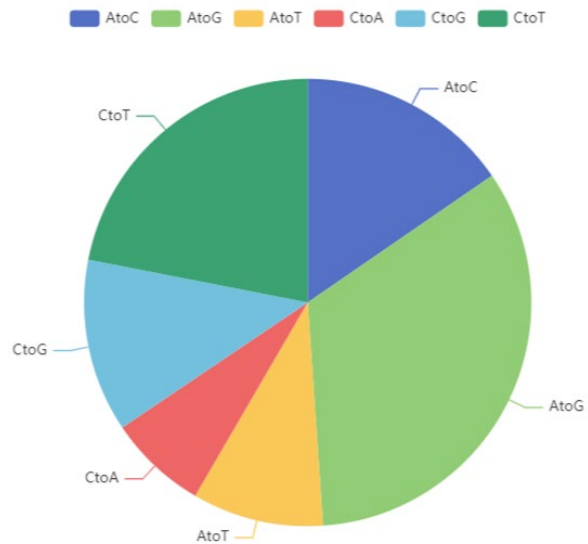
Visualize variantsfile

Data Import

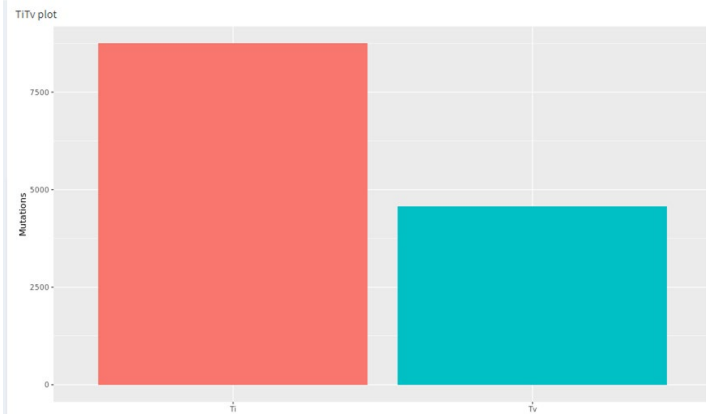
SNP Data

Amino Acid Changes

SNPs types Pie plot pie of SNPs types

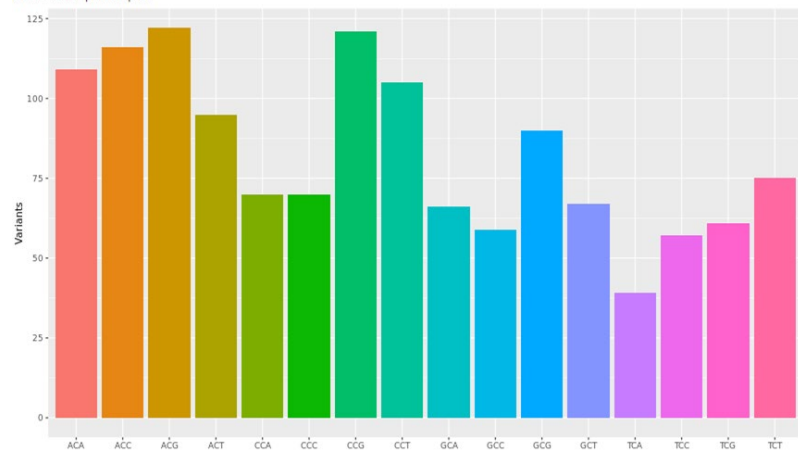


TiTv distribution TiTv plot

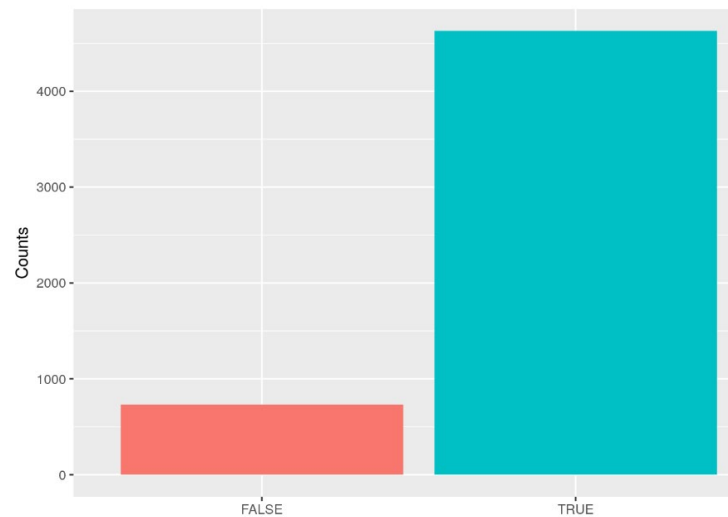




Trinucleotide pattern plot Trinucleotide pattern plot

Trinucleotide pattern plot

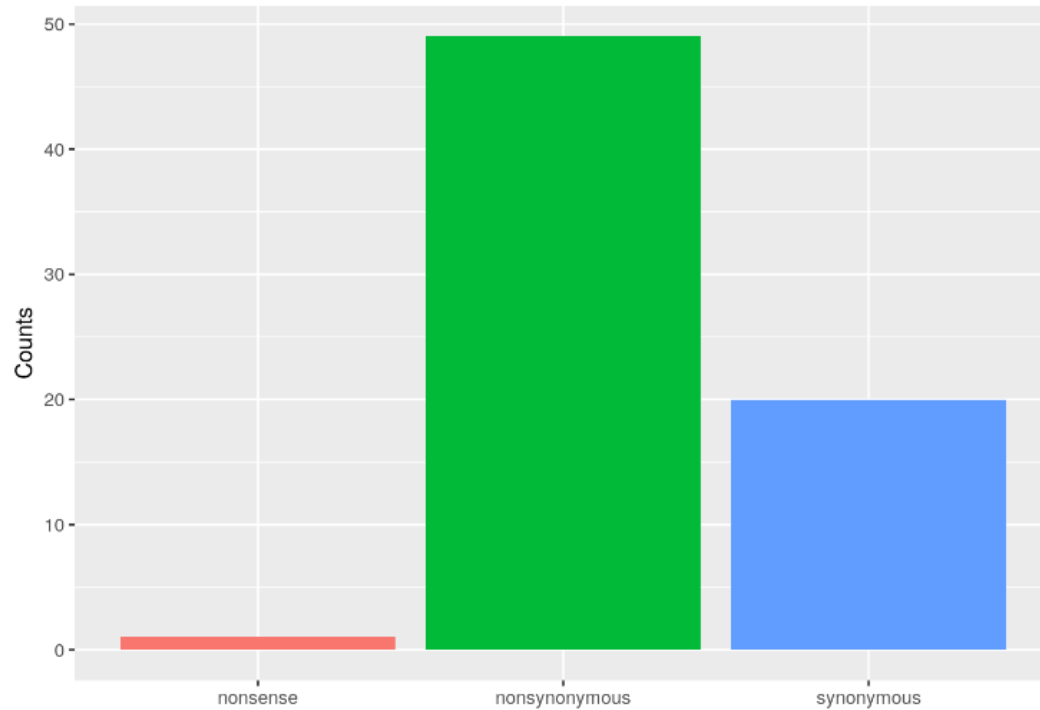


True and False variants count



 Data Import SNP Data Amino Acid Changes

### The Amino Acid Changes





# **CNVviz App**

**For CNV data visualization**

# 3. CNVviz

CNVviz



- Import Data
- Recurrent Regions
- Overlap Analysis
- Overlap Permutation Test

## Visualize CNVs data

### Import File

Import the variant data csv file here

Upload csv file

Browse...

Copy Number Calls.csv

Upload complete

☒ Demo

Input file

Show 10 entries

Search:

	chr	start	end	NE_id	state
1	chr1	16947	45013	NE001423	3
2	chr1	36337	67130	NE001426	3
3	chr1	16947	36337	NE001428	3
4	chr1	36337	105963	NE001519	3
5	chr1	36337	83412	NE001534	3
6	chr1	36337	83412	NE001648	3
7	chr1	36337	83412	NE001722	3
8	chr1	36337	89725	NE001741	3
9	chr1	36337	83412	NE001743	3
10	chr1	16947	89725	NE001753	3

Showing 1 to 10 of 3,000 entries

Previous 1 2 3 4 5 ... 300 Next

- Import Data
- Recurrent Regions
- Overlap Analysis
- Overlap Permutation Test

## Identifying recurrent regions

### About

The The GISTIC method is used to identify those regions of the genome that are aberrant more often than would be expected by chance, with greater weight given to high amplitude events (high-level copy-number gains or homozygous deletions) that are less likely to represent random aberrations (Beroukhim et al., 2007).

Please define the significance threshold to filter recurrent CNVs.

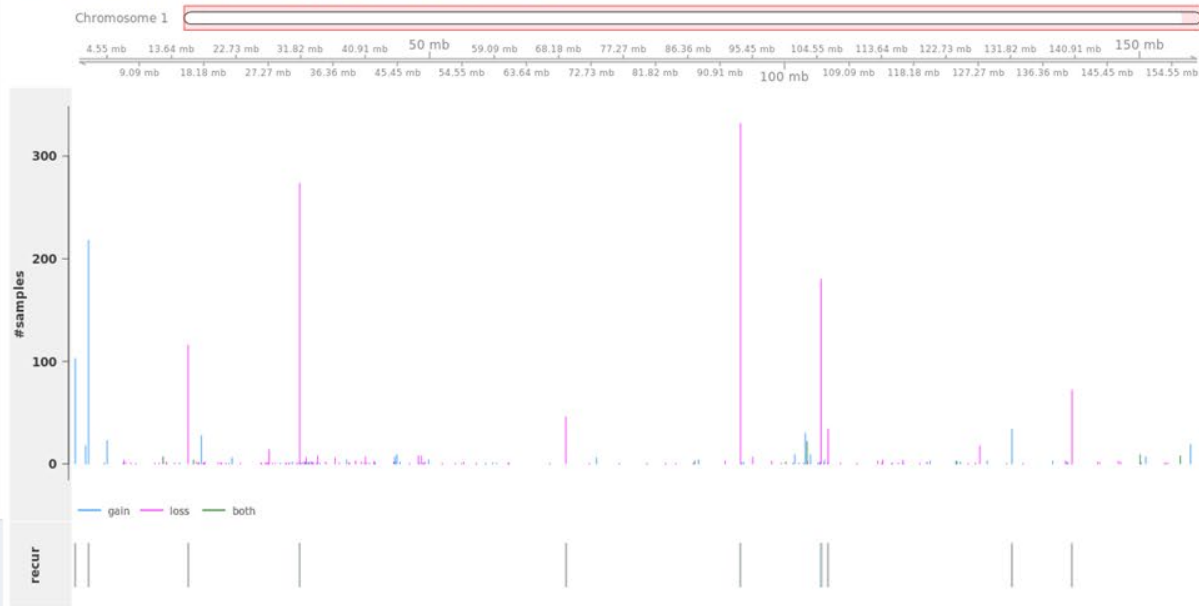
### P value

Please define the chromosome

### Chromosome

### Recurrent

The plot summarize CNV regions, a valid UCSC genome assembly, and a chromosome of interest.



- Import Data
- Recurrent Regions
- Overlap Analysis
- Overlap Permutation Test

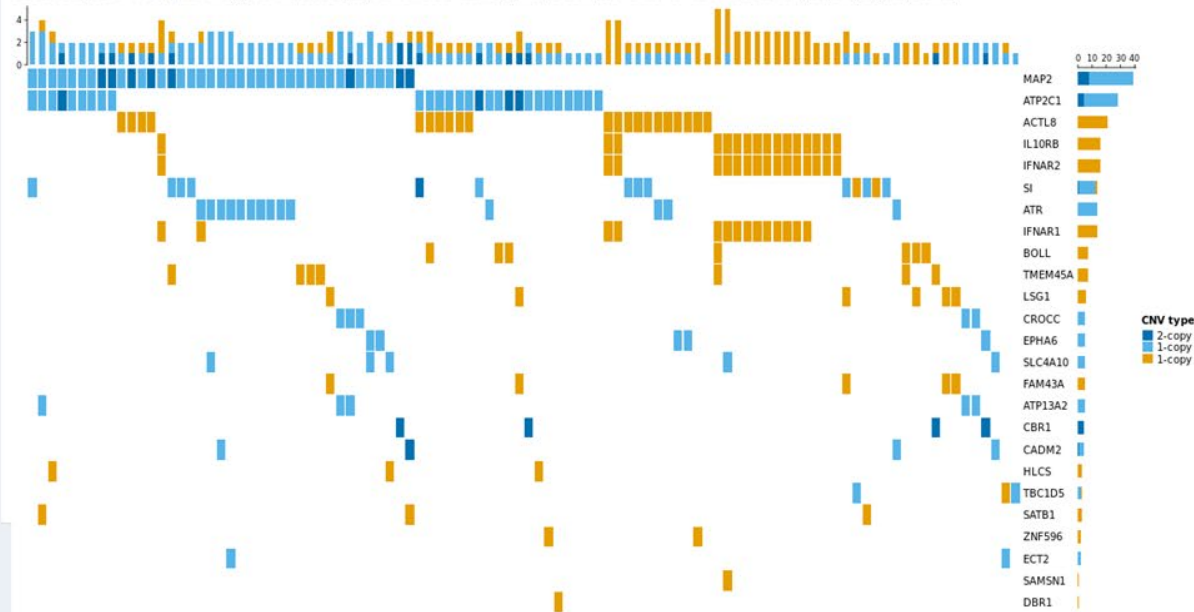
## Overlap analysis of CNVs with functional genomic regions

### About

It is of interest whether the resulting CNV regions overlap with functional genomic regions such as genes, promoters, or enhancers.

### Overlap Plot

Stacked barplots on the top and the right of the plot display the number of altered genes per sample and the number of altered samples per gene, respectively.



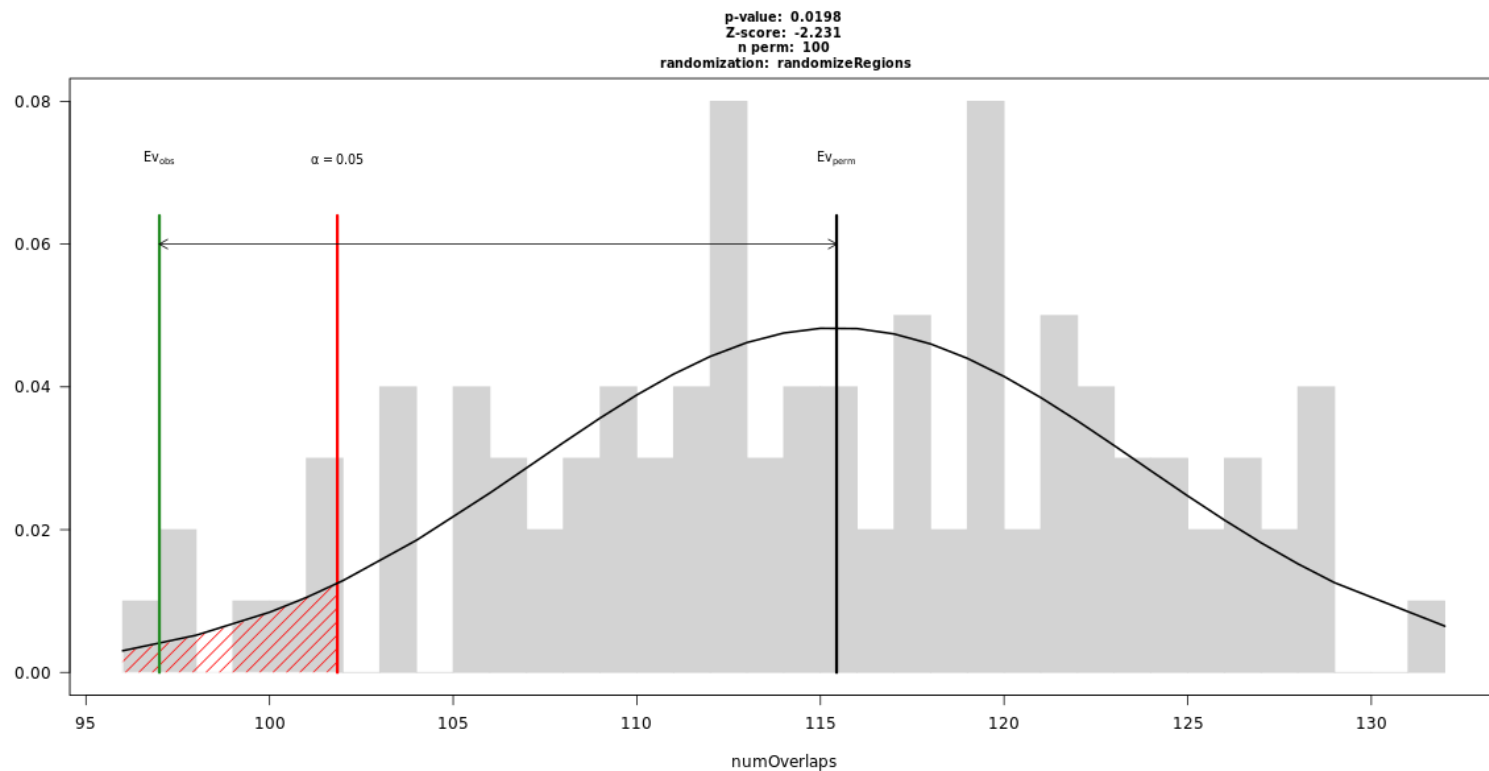
OncoPrint plot

= illustrate the original CNV calls on overlapping protein-coding genes

- Import Data
- Recurrent Regions
- Overlap Analysis
- Overlap Permutation Test

## Overlap permutation test

Overlap Plot



## Acknowledgements



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