# 1 Search by Gene

## General Instruction

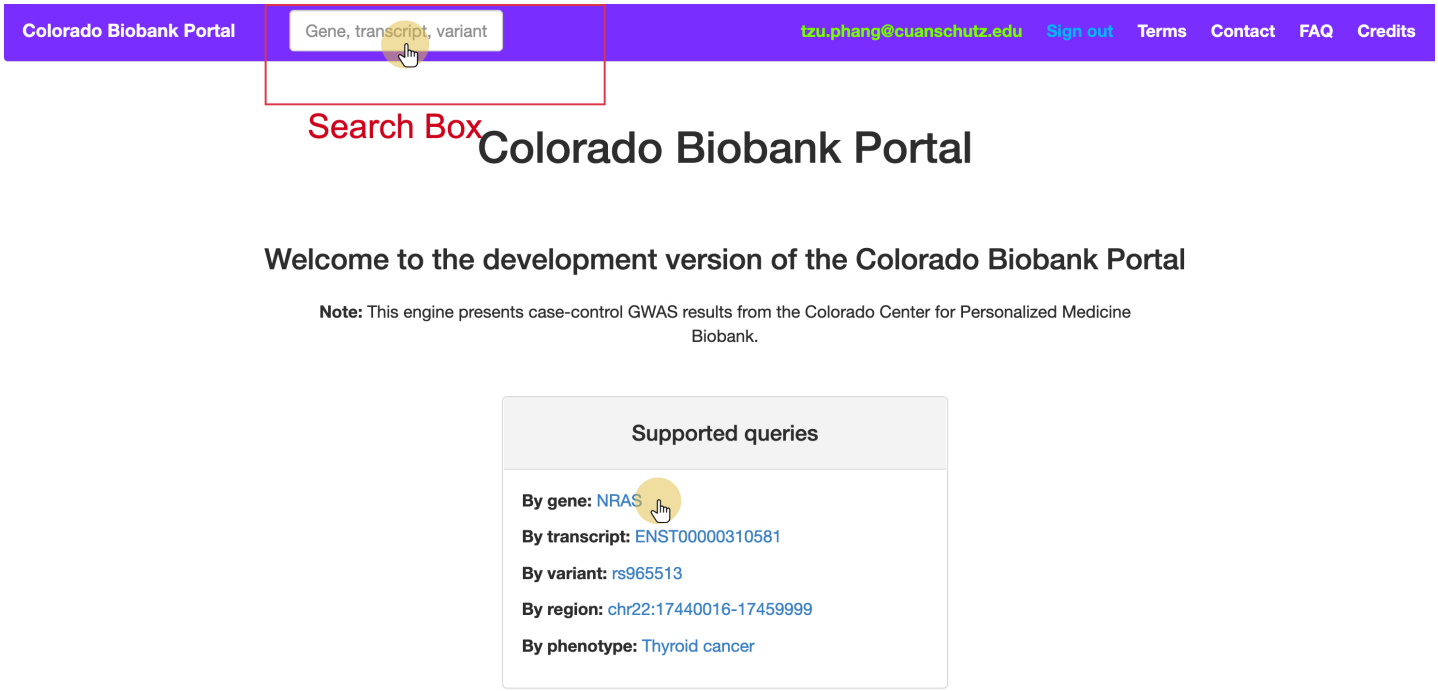
You can search the Colorado Biobank Portal (CBP) using your favorite gene.

The easiest way to try this out to **Search by gene**, click the **NRAS** link

# 2 Insert Gene Symbols

## General Instruction

Alternatively, you can type the **Gene Symbols** in the **Search Box** blank space.



# 3 Gene Symbol Suggestion

**General Instruction**

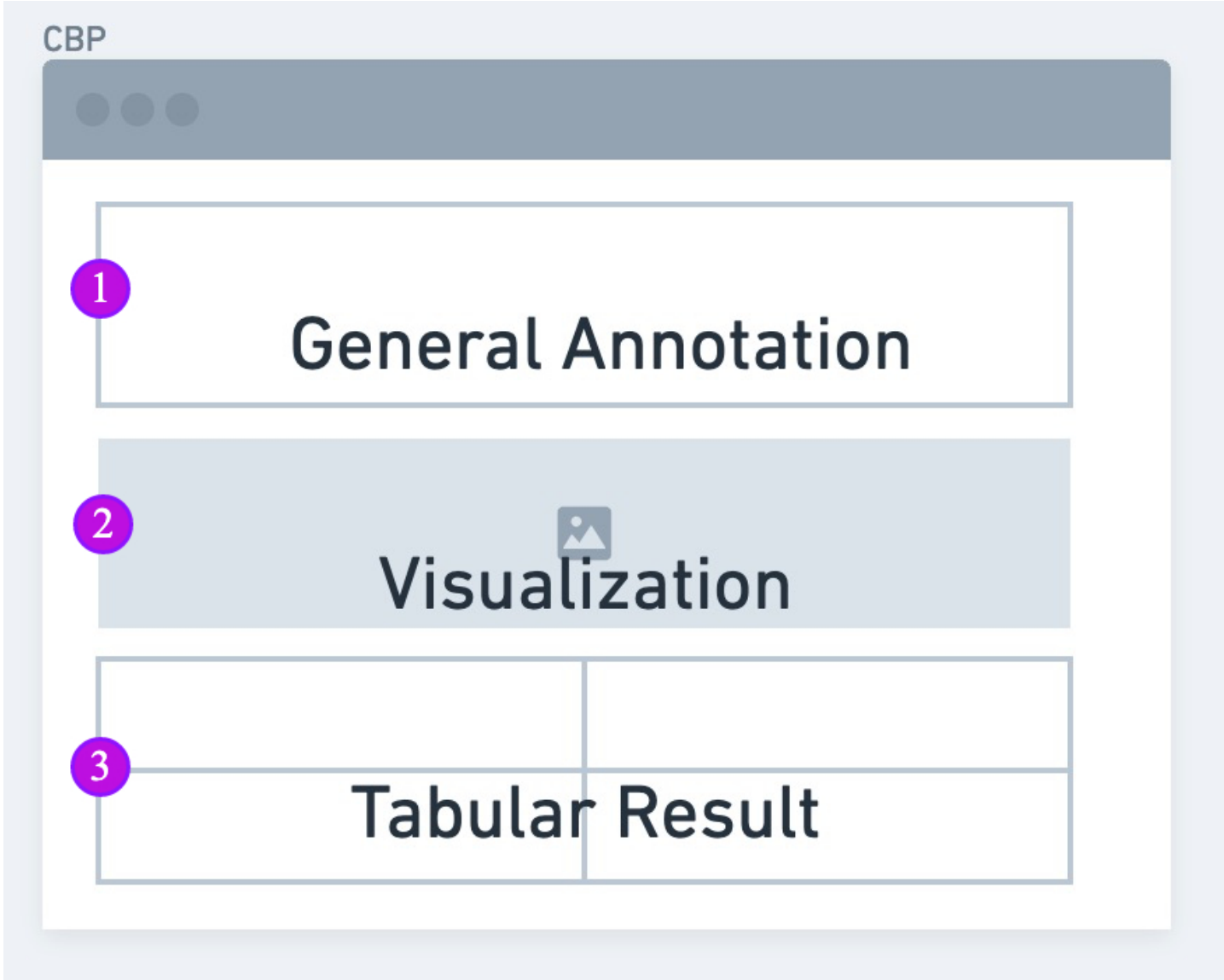
While you type, an auto-complete feature will take over to match your Gene Symbol name. Please select one of the entry from the auto-complete list.

# 4 Results Layout

**General Instruction**

The search result will be displayed in this general layout that consists of three segments:

1. General Annotation: information on the search results and relevant external links
2. Visualization: graphical representation of the search results
3. Tabular Result: table layout of search results summary statistics.

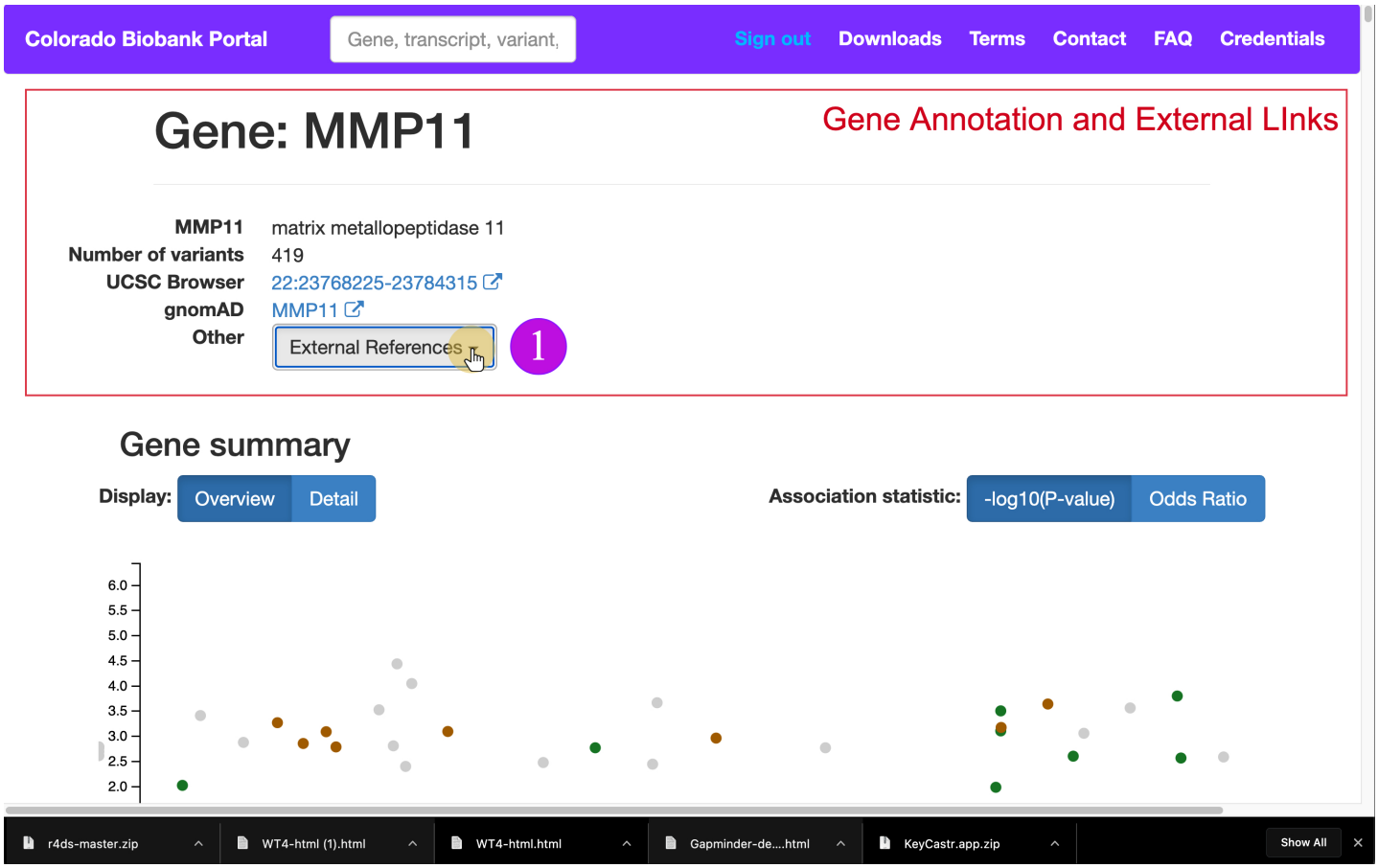


# 5 General Annotation

**General Instruction**

The top segment of the result page lists general gene info and external links pertaining to the gene

1. Click on the **External References** to reveal more external links option



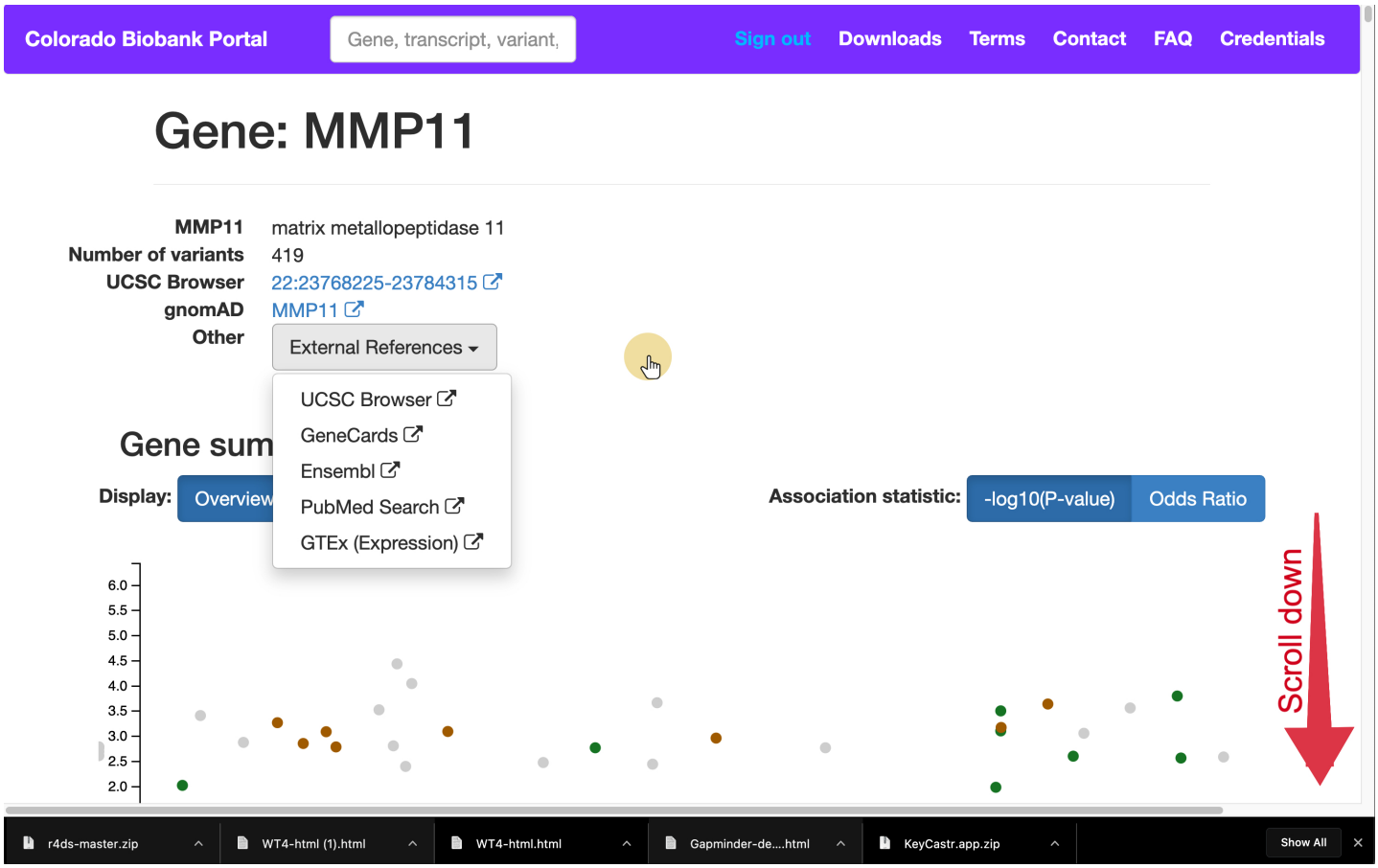
# 6 More external link

**General Instruction**

More external links pertainig to the gene can be found under the pull down manu

**Scroll for more ...**

Scroll down to see more result

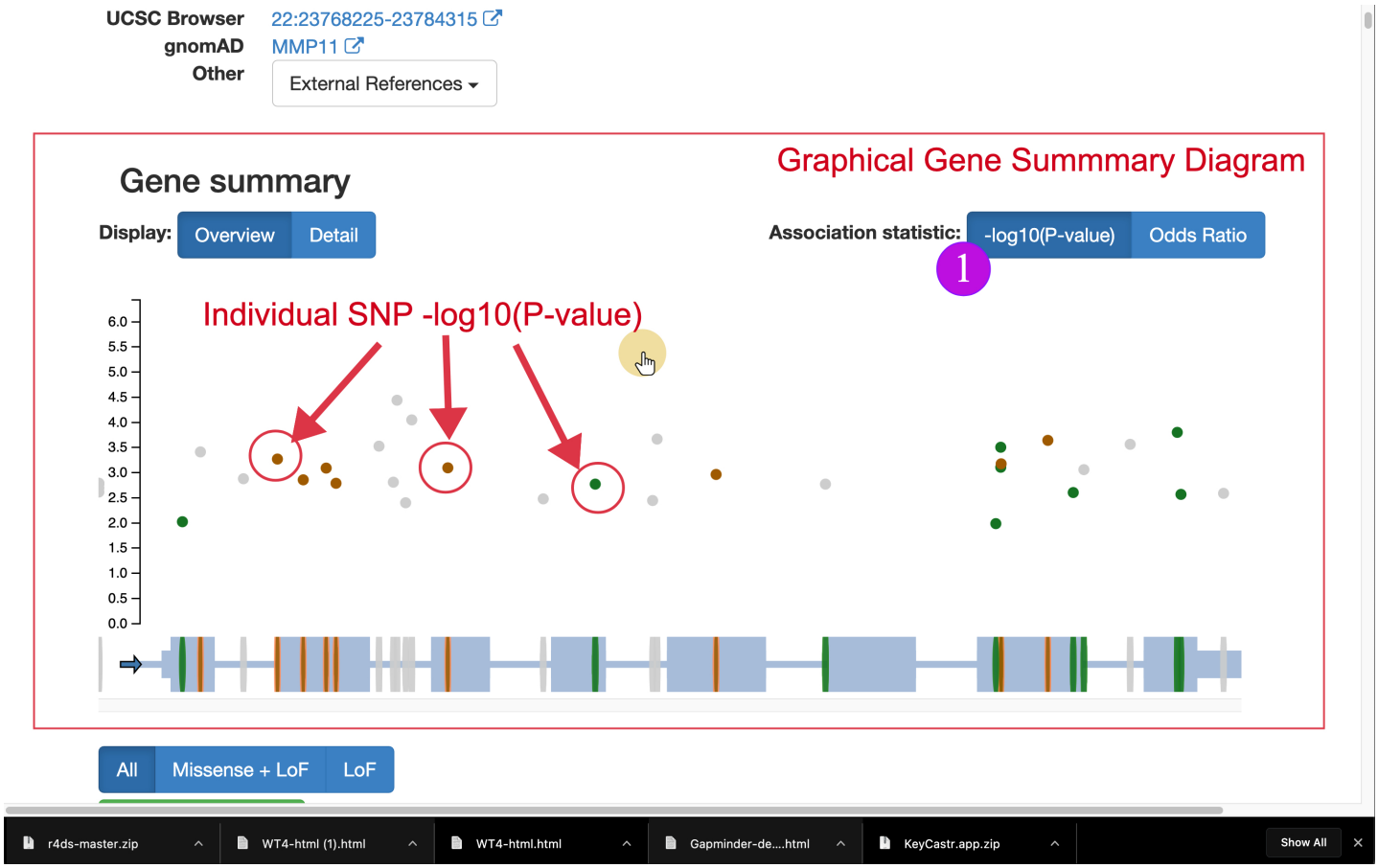


# 7 Visualization: -log(p-value)

**General Instruction**

This segment reveal the graphical representation of the gene of interest.

1. By default, each dot represents a SNP in -log(p-value). You can also click on the "-log10(P-value)" box to activate it.



# 8 Visualization: Odds Ratios

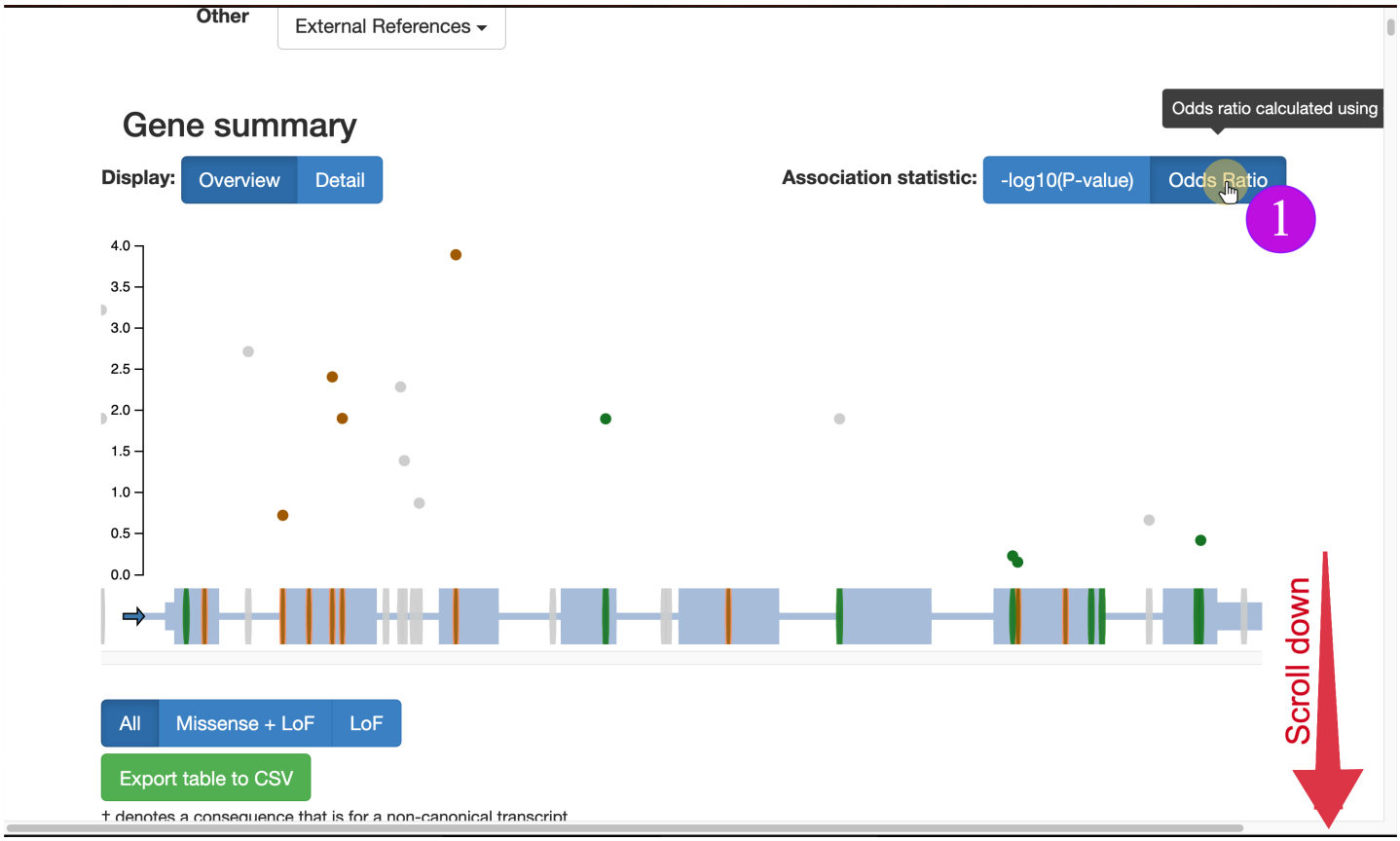
## General Instruction

You can also view the result in "Odds Ratios" unit value

1. Click on the "Odds Ratio" box will activate this feature too.

## Scroll for more

Scroll down to reveal more result



# 9 Tabular Summary Statistics

**General Instruction**

The table shows summary statisticss for all the SNPs

1. **Variant**: Single Nucleotide Polymorphism (SNP), labeled with these genetic info:
    a. Chromosome location (here: Chromosome 22, position 23,763,464)
    b. Genetic alteration (here: altered from nucleotide G to A)
    c. rsID (here: rs575501011)
2. **Position**: SNP genomic coordinate position for hg38
3. **Gene Symbol**: official Gene Symbol
4. **Consequence**: the repercussion of the genetic alteration (mutation) which could be either benign or damaging
5. **Annotation**: additional information about the SNP as it relates to gene(s)
6. **Minor AF**: Minor Allele Frequency: the frequency at which the second most common allele occurs in a given population (here it is based on CCPM Biobank samples)
7. **Phenotype**: the phenotype used to calculate summary statistics
8. **Odds Ratio**: the ratio of the odds of phenotype (disease) among the exposed to the odds of phenotype among the unexposed.
9. **P-value**: Genome Wide Association Study (GWAS) analysis based on the phenotype which tells us how likely a putative phenotype (disease) associated variant is due to random chance

† de a consequence that is for a non-canon nscrip

| 1 Variant | 2 Position | 3 Gene symbol | 4 Consequence | 5 Annotation | 6 Minor AF | 7 Phenotype | 8 Odds Ratio | 9 P-value |
|---|---|---|---|---|---|---|---|---|
| 22:23763464 G / T (rs575501011) | 23763464 | MMP11 | | upstream gene | 0.000691584 | Intracranial hemorrhage (injury) | 19.7 | 0.00140 |
| 22:23763602 G / A (rs73396542) | 23763602 | MMP11 | | upstream gene | 0.194672 | Iron deficiency anemia secondary to blood loss ... | 1.25 | 0.00190 |
| 22:23763630 G / A (rs114418367) | 23763630 | MMP11 | | upstream gene | 0.000861755 | Malignant neoplasm of ovary | 22.7 | 0.00147 |
| 22:23763660 A / C (rs866456155) | 23763660 | MMP11 | | upstream gene | | Leukemia | 10.7 | 0.00107 |
| 22:23763748 G / A (rs906072089) | 23763748 | MMP11 | | upstream gene | | Rheumatic disease of the heart valves | 23.5 | 0.00169 |
| 22:23763821 G / C (rs179465) | 23763821 | MMP11 | | upstream gene | 0.0916575 | Concussion | 0.303 | 0.000118 |
| 22:23763985 G / A (rs2298374) | 23763985 | MMP11 | | upstream gene | | Skin cancer | 29.6 | 0.00247 |

# 10 Tabular Summary Statistics: Consequence Example

## General Instruction

An example of consequence output

All | Missense + LoF | LoF

Export table to CSV

† denotes a consequence that is for a non-canonical transcript

| Variant | Position | Gene symbol | Consequence | Annotation | Minor AF | Phenotype | Odds Ratio | P-value |
|---|---|---|---|---|---|---|---|---|
| 1:114708192 T / C (rs909455480) | 114708192 | NRAS | c.*5A>G | splice region | | Myopia | 14.6 | 0.00712 |
| 1:114713865 G / A (rs142739534) | 114713865 | NRAS | p.Gly75/189Gly | synonymous | | Other and unspecified complications of birth, p... | 31.9 | 0.00301 |
| 1:114709659 C / T (rs143020946) | 114709659 | NRAS | p.Leu120/189Leu | synonymous | 0.000645953 | Dysmetabolic syndrome X | 7.62 | 0.00371 |
| 1:114708552 G / A (rs374061873&COSV65739652) | 114708552 | NRAS | p.Pro185Ser | missense | | Early or threatened labor, hemorrhage in early ... | 22.0 | 0.00255 |
| 1:114705879 CT / C (rs770180355) | 114705879 | NRAS | | 3' UTR | | Acute bronchitis and bronchiolitis | 53.7 | 0.00380 |
| 1:114704375 A / C (rs886259422) | 114704375 | NRAS | | downstream gene | | Abnormal findings on mammogram or breast exam | 9.36 | 0.00192 |
| 1:114706928 A / C (rs774604143) | 114706928 | NRAS | | 3' UTR | | Other immunological findings | 41.6 | 0.00727 |
| 1:114707511 A / C (rs555171083) | 114707511 | NRAS | | 3' UTR | | Psychogenic and somatoform disorders | 37.9 | 0.00124 |
| 1:114714742 A / G (rs9724624) | 114714742 | NRAS | | intron | 0.0433714 | Nontoxic multinodular goiter | 1.58 | 0.00122 |
| 1:114712736 A / T (rs573797597) | 114712736 | NRAS | | intron | 0.000312664 | Other disorders of thyroid | 8.57 | 0.00220 |
| 1:114708093 A / T (rs769896264) | 114708093 | NRAS | | intron | | Influenza | 51.2 | 0.00434 |
| 1:114716848 A / T (rs2273267) | 114716848 | NRAS | | upstream gene | 0.00909405 | Hypotension NOS | 2.92 | 0.00261 |
| 1:114708698 C / T (rs9724641) | 114708698 | NRAS | | intron | 0.0089222 | Schizophrenia and other psychotic disorders | 2.59 | 0.00184 |
| 1:114709234 C / T (rs1016348904) | 114709234 | NRAS | | intron | | Gram negative septicemia | 77.4 | 0.00224 |
| 1:114709312 G / A (rs115405784) | 114709312 | NRAS | | intron | 0.000340101 | Mood disorders | 0.126 | 0.00194 |
| 1:114704595 G / A (rs147926293) | 114704595 | NRAS | | 3' UTR | 0.00114319 | Disturbances of sensation of smell and taste | 8.85 | 0.000817 |
| 1:114713267 G / A (rs975060114) | 114713267 | NRAS | | intron | | Congestive heart failure, nonhypertensive | 14.7 | 0.000710 |