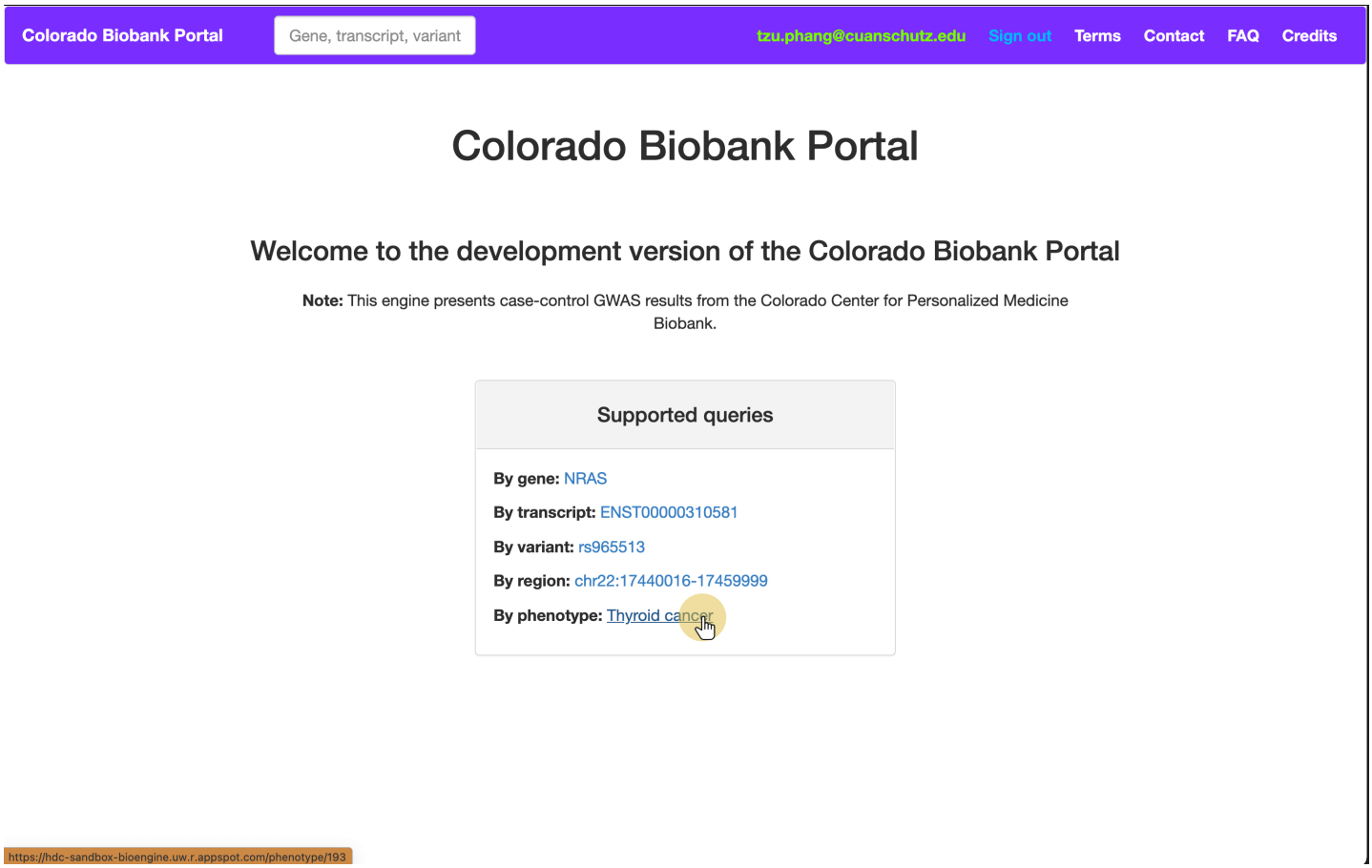
1 Search by Phenotype

General Instruction

You can search Colorado Biobank Portal (CBP) using your favorite phenotype

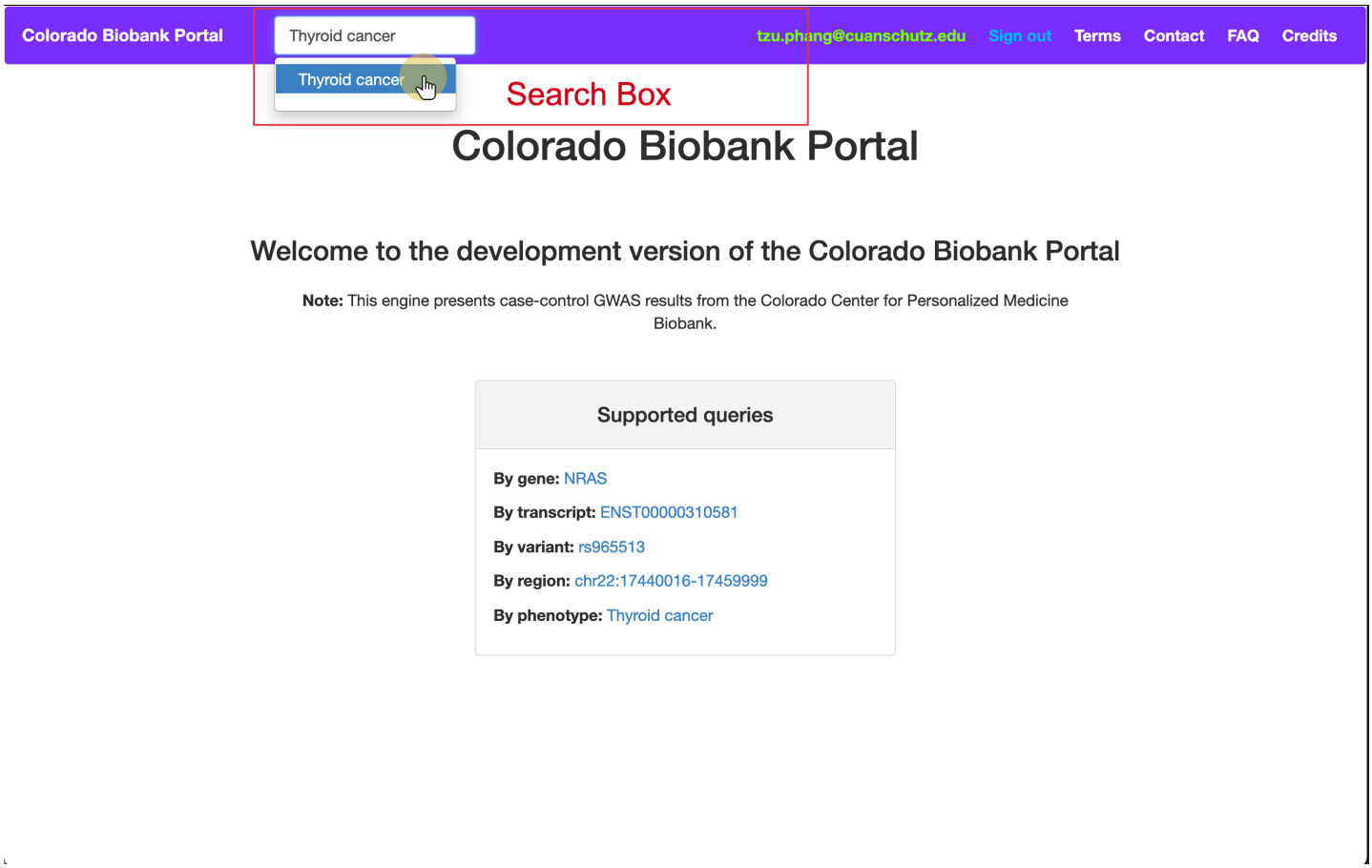
The easiest way to try this out is to click on search **By phenotype**



2 Insert Phenotype

General Instruction
Alternatively, you can type the **Phenotype** in the **Search Box** blank space

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

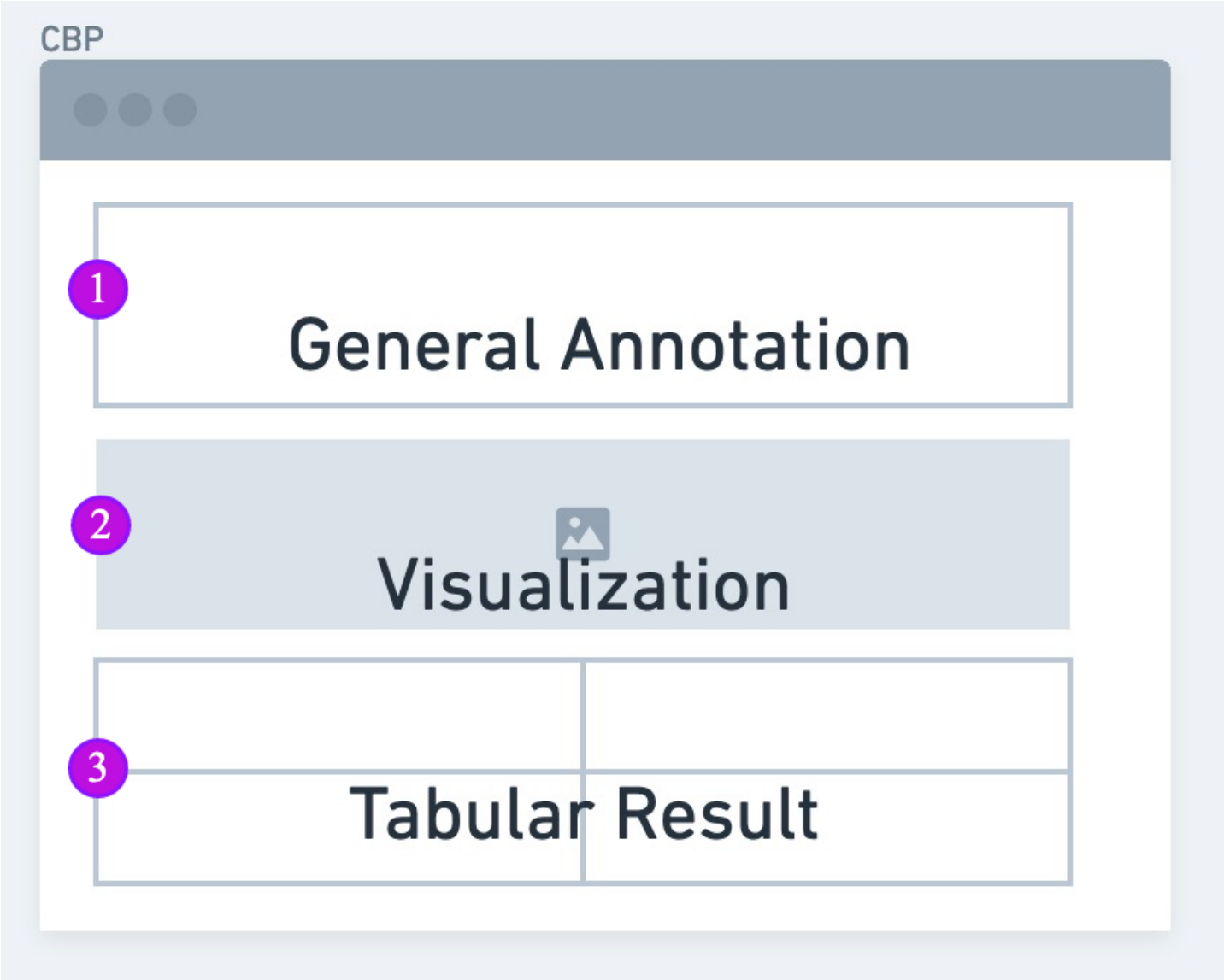


3 Results Layout

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.

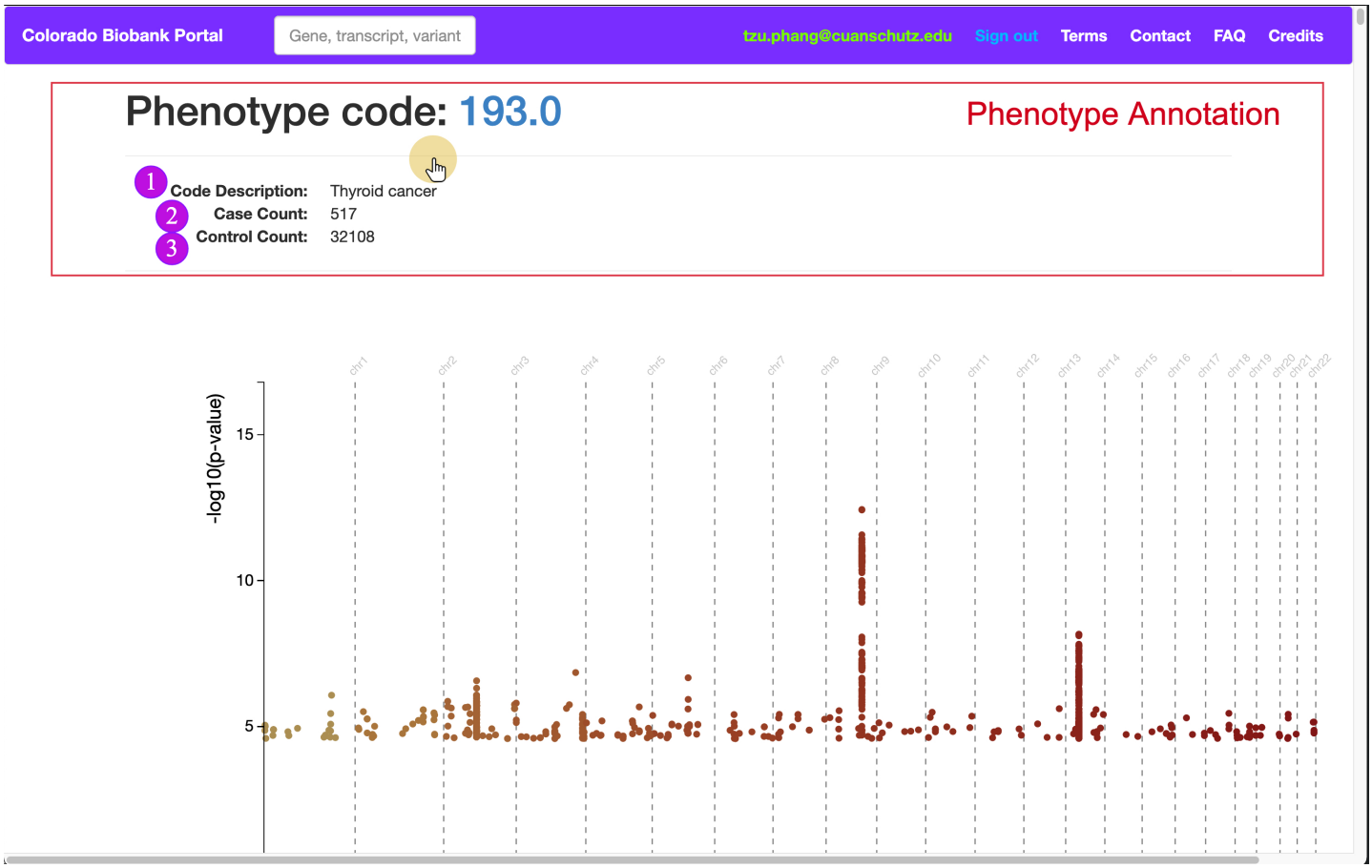


4 General Annotation

General Instruction

The top segment of the result page lists general info for the phenotype and some summary statistics:

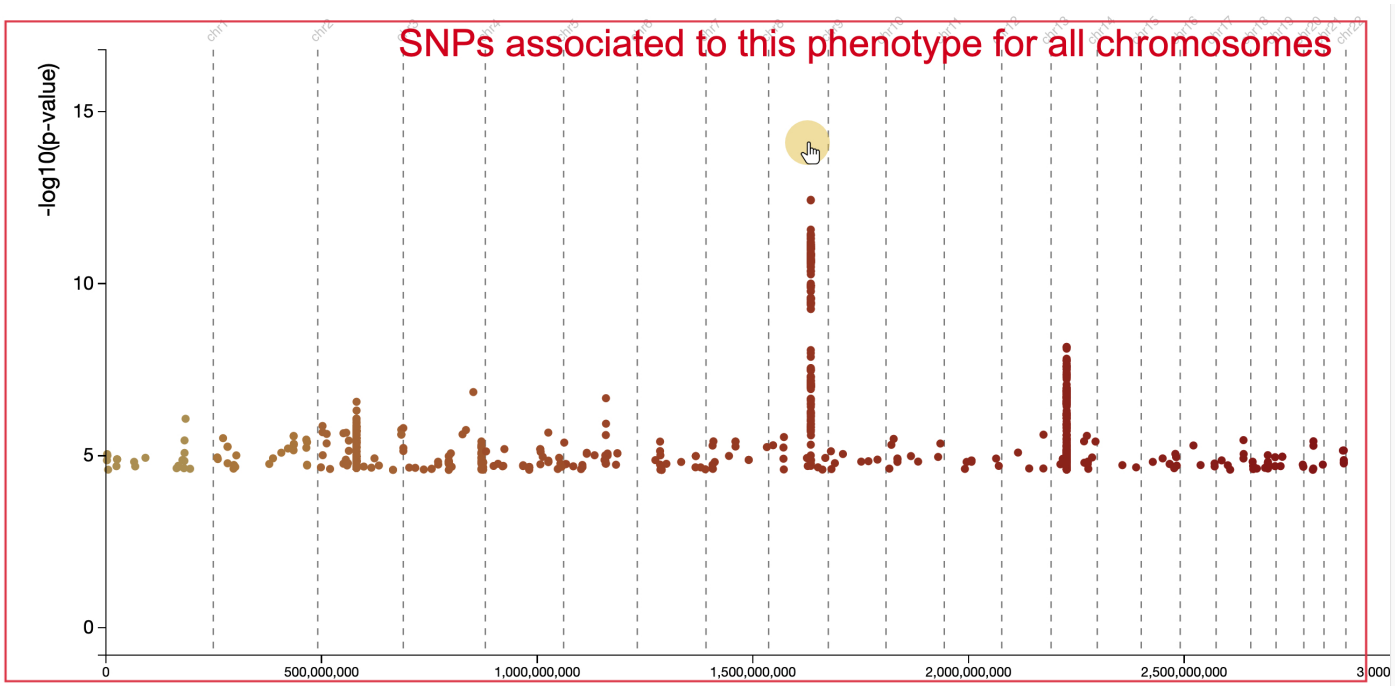
1. Phenotype
2. Case count on the biobank population
3. Control count on the biobank population



5 Visualization

General Instruction

This segment reveal the graphical representation of PheWAS analysis of all SNP pertaining to this phenotype for all chromosomes.



Note: 1000 variants with lowest p-value are shown.

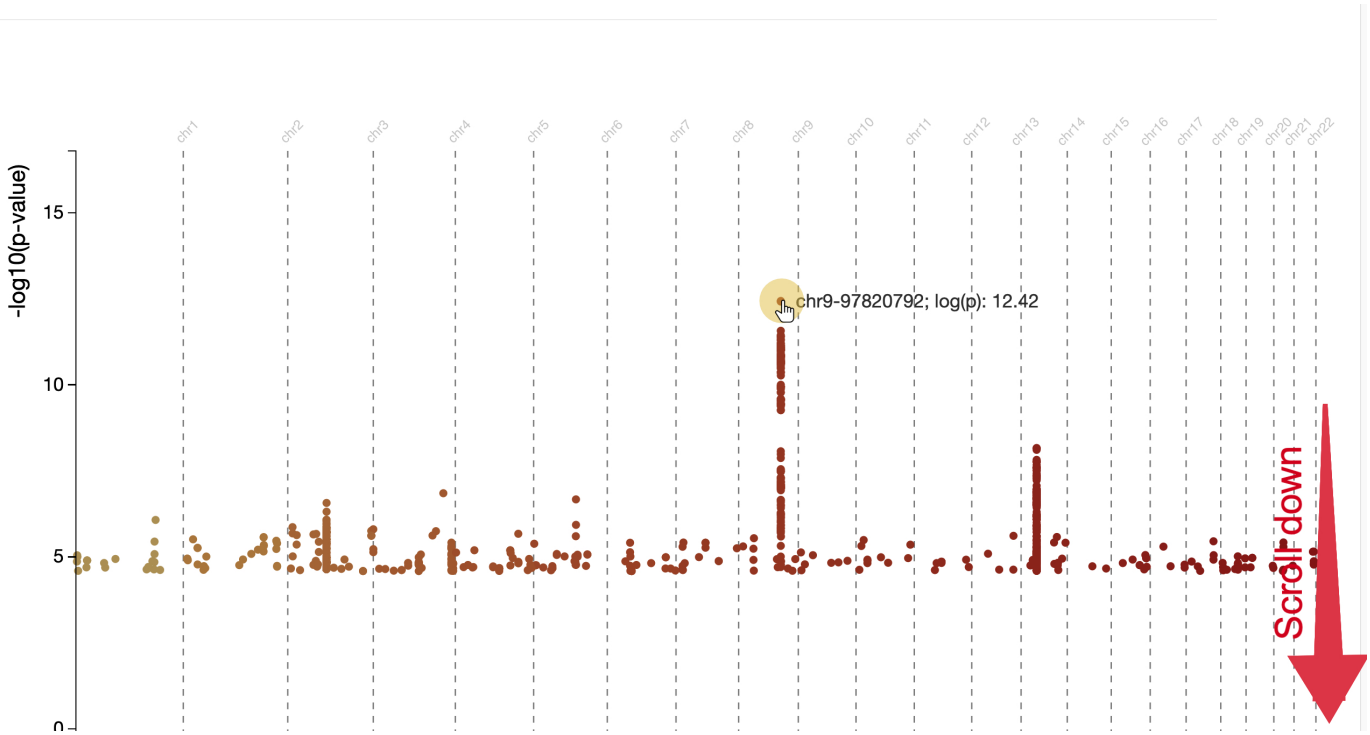
6 Visualization

General Instruction

Mouse over the dot to reveal SNP summary statistics

Scroll for more

Scroll down to reveal more result



7 Tabular Summary Statistics

General Instruction

The table shows summary statisticss for the top 1000 most significant SNPs associated with the phenotype of interest:

1. **Variant**: Single Nucleotide Polymorphism (SNP), labeled with these genetic info:
    a. Chromosome location (here: Chromosome 4, position 72,618)
    b. Genetic alteration (here: altered from nucleotide T to C)
    c. rsID (here: rs11734652)
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. **Odds Ratio**: the ratio of the odds of phenotype (disease) among the exposed to the odds of phenotype among the unexposed.
8. **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

All | Missense + LoF | LoF

1 table to CSV

Variant	Position	Gene symbol	Consequence	Annotation	AF	Odds Ratio	P-value
4:72618 T / C (rs11734652)	72618	ZNF595		intron	0.0511955	0.432	0.00000158
4:177452 G / T (rs11728456)	177452	ZNF718		intron	0.0523924	0.463	0.00000735
4:197698 A / G (rs77472447)	197698	AC108475.1 , ZNF718		intron	0.0539964	0.465	0.00000600
6:1462108 C / T (rs937952912)	1462108			intron		40.3	0.00000418
5:1767672 C / T (rs116185244)	1767672			intergenic	0.001046	17.9	0.00000753
1:2416601 T / C (rs10910068)	2416601	PEX10		intron	0.349851	1.33	0.0000137
1:2875882 G / T (rs1037644371)	2875882			regulatory region		62.9	0.00000904
1:3539984 C / A (rs940128132)	3539984	MEGF6		intron		49.7	0.00000996
6:4271400 G / A (rs1014074434)	4271400			regulatory region		32.1	0.0000176
19:4339747 A / G (rs11669563)	4339747	MPND , STAP2 , AC104521.1		intron	0.00174261	6.58	0.0000150
1:5088245 G / A (rs531117898)	5088245	LINC02782	n.338+6C>T	splice region		134	0.0000256
19:5324099 C / T (rs145930256)	5324099	PTPRS		intron	0.000758819	12.9	0.0000200
19:5794278 C / T (rs924048234)	5794278	DUS3L		upstream gene	0.000368769	25.2	0.0000246
6:5810797 G / A (rs999183570)	5810797	FARS2		intron		149	0.0000176
10:6070328 C / T (rs79961688)	6070328	RPL32P23		downstream gene	0.00058887	19.1	0.00000746
3:7297964 C / T (rs536574194)	7297964	GRM7		intron		55.3	0.0000219
10:7322125 TA / T (rs756868278)	7322125	SFMBT2		intron		173	0.0000247
11:7792533 A / C (rs765168193)	7792533	OR5P2 , AC044810.3 , AC044810.2		intron		103	0.0000239
2:9254800 G / A (rs148667240)	9254800	ASAP2		intron	0.00049108	31.3	0.0000113
2:9257326 A / G (rs137905963)	9257326	ASAP2		intron	0.00049108	31.3	0.0000113
2:9264851 A / C (rs143875080&COSV55626602)	9264851	ASAP2		intron	0.000502311	31.0	0.0000117

8 Tabular Summary Statistics: Consequence Example

General Instruction

An example of consequence output

All | Missense + LoF | LoF

Export table to CSV

Variant	Position	Gene symbol	Consequence	Annotation	AF	Odds Ratio	P-value
19:39875779 G / A (rs377299687)	39875779	FCGBP	p.Tyr3417/4217Tyr	synonymous		43.5	0.0000237
9:97854739 T / C (rs3021526)	97854739	FOXE1 , PTCSC2	p.Ser275/373Ser	synonymous	0.380799	1.49	2.94e-10
12:120216438 G / C (rs140854897)	120216438	PXN , PXN-AS1 , AC004263.2	p.Pro107/207Pro	synonymous	0.00293749	4.95	0.0000122
12:51820862 G / A (rs749758515)	51820862	AC068987.3 , FIGNL2 , AC068987.2	p.Leu518/653Leu	synonymous		57.9	0.0000152
9:97854301 C / T (rs3021523)	97854301	FOXE1 , PTCSC2	p.Leu129/373Leu	synonymous	0.241562	1.45	1.00e-7
11:17772627 C / T (rs139606715)	17772627	KCNC1	p.Ile511/511Ile	synonymous		31.5	0.00000326
6:43504970 T / C (rs3734689)	43504970	TJAP1 , LRRC73 , POLR1C	p.His263/557His	synonymous	0.0289676	0.313	0.0000217
1:5088245 G / A (rs531117898)	5088245	LINC02782	n.338+6C>T	splice region		134	0.0000256
9:97896500 C / CTTAT (rs35588227)	97896500			non coding transcript exon	0.342742	1.54	1.48e-11
9:97771035 A / ACT (rs5899324)	97771035	PTCSC2		intron	0.3744	1.53	1.82e-11
9:97879489 C / CCACCA (rs138322543)	97879489			intergenic	0.347513	1.53	1.97e-11
9:97869517 C / CCAAA (rs10555589)	97869517			regulatory region	0.346787	1.53	2.20e-11
9:97871868 C / CAG (rs35660240)	97871868			regulatory region	0.346897	1.53	2.27e-11
9:97856808 T / TCTC (rs3029497)	97856808	FOXE1 , PTCSC2		upstream gene	0.379951	1.49	3.40e-10
14:36052840 C / CG (rs35238964)	36052840	ILF2P2 , AL133304.3		intron	0.468735	0.688	7.77e-9
14:36050489 C / CA (rs35474452)	36050489	ILF2P2 , AL133304.3		intron	0.471741	0.698	2.75e-8
9:97834407 T / TA (rs35103839)	97834407	PTCSC2		intron	0.243287	1.47	2.84e-8
14:36084259 G / GAAGT (rs71124738)	36084259	LINC00609		intron	0.480253	0.702	4.44e-8
9:97882563 T / TA (rs11307149)	97882563			intergenic	0.241026	1.46	6.87e-8
9:97836560 C / CT (rs34172707)	97836560	PTCSC2		intron	0.275647	1.40	9.96e-7