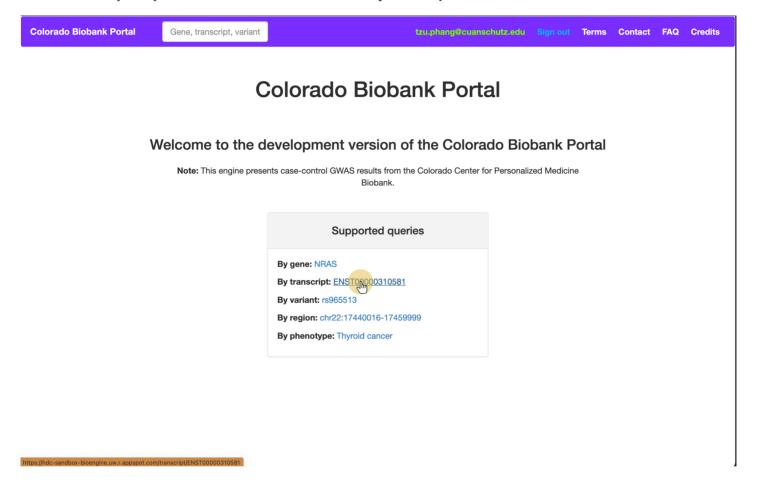# 1 Search by Transcript

General Instruction

You can search the Colorado Biobank Portal (CBP) using your favorate gene transcript ID.

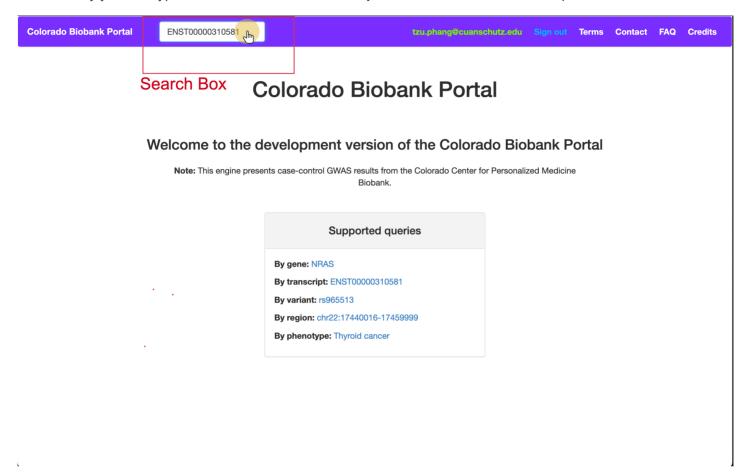The easiest way to try this out is to click on the Search **by transcript** link

---

Colorado Biobank Portal   Gene, transcript, variant   **tzu.phang@cuanschutz.edu** Sign out **Terms** **Contact** **FAQ** **Credits**

Colorado Biobank Portal

Welcome to the development version of the Colorado Biobank Portal

Note: This engine presents case-control GWAS results from the Colorado Center for Personalized Medicine Biobank.

Supported queries

By gene: NRAS

By transcript: ENST00000310581

By variant: rs965513

By region: chr22:17440016-17459999

By phenotype: Thyroid cancer

---

https://hdc-sandbox-bioengine.uw.r.appspot.com/transcript/ENST00000310581

2 Insert Transcript ID

General Instruction

Alternatively you can type the **ENSEMBL Gene Transcript ID** in the **Search Box** blank space
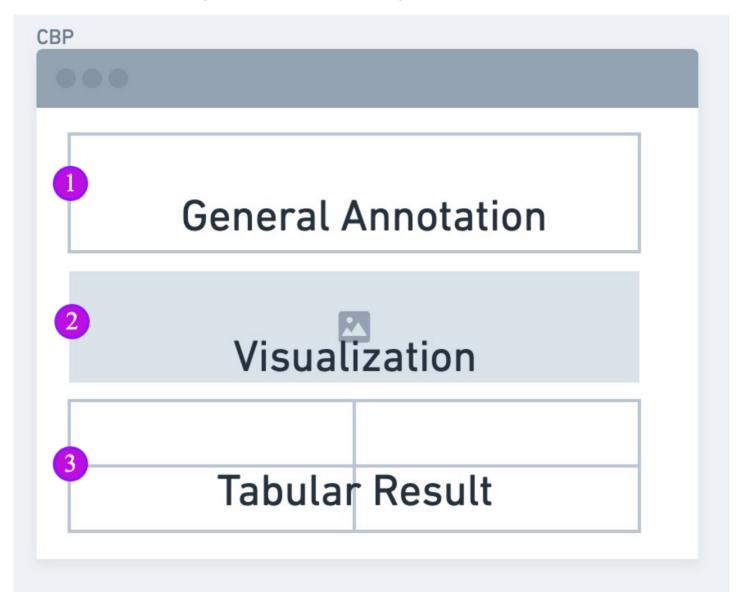
3 Results Layout

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

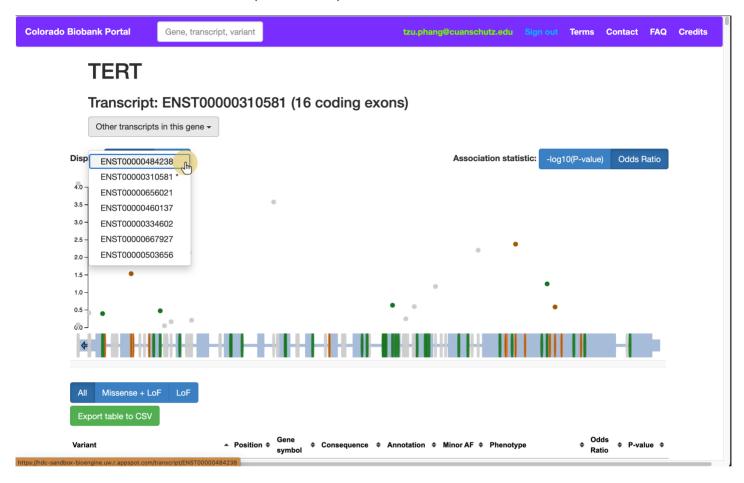The top segment of the result page lists general transcript info

5 Search Alternative Transcript

General Instruction

You can also select alternative trancripts from the pull down selection.

| Colorado Biobank Portal | Gene, transcript, variant | tzu.phang@cuanschutz.edu | Sign out | Terms | Contact | FAQ | Credits |

TERT

Transcript: ENST00000310581 (16 coding exons)

Other transcripts in this gene ▾

ENST00000484238
ENST00000310581 *
ENST00000656021
ENST00000460137
ENST00000334602
ENST00000667927
ENST00000503656

Association statistic: -log10(P-value) | Odds Ratio

All | Missense + LoF | LoF

Export table to CSV

| Variant | | Position | Gene symbol | | Consequence | | Annotation | | Minor AF | Phenotype | | Odds Ratio | | P-value | |

https://hdc-sandbox-bioengine.uw.r.appspot.com/transcript/ENST00000484238
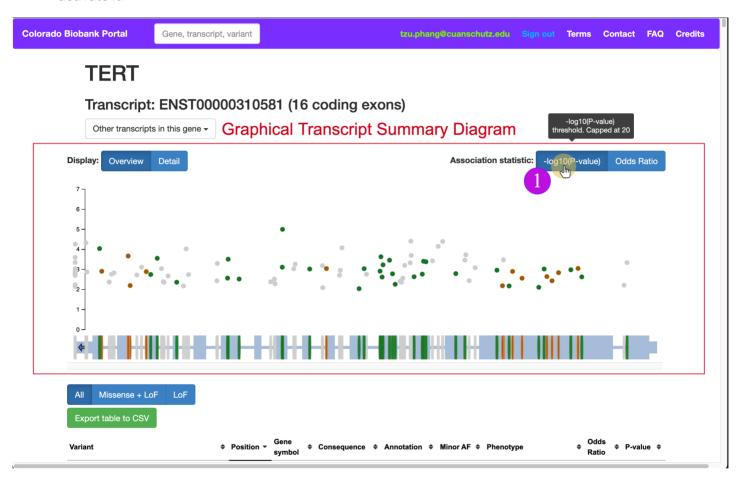
6 Visualization: -log(p-value)

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.
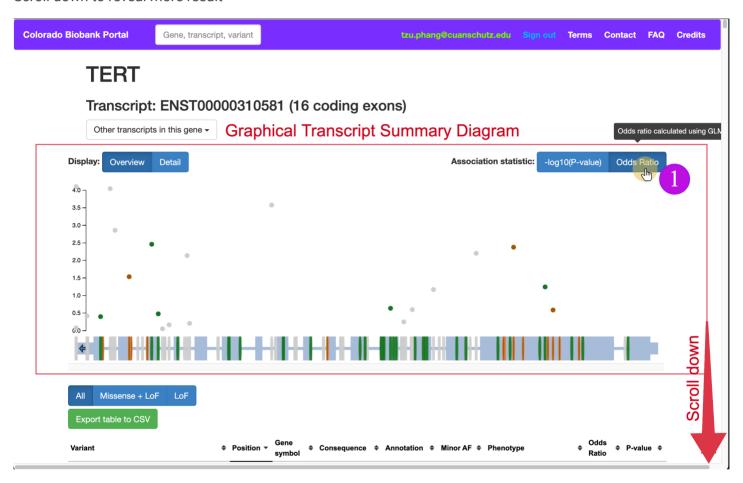
7 Visualization: Odds Ratios

General Instruction
You can also view the result in "Odds Ratio" unit value

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

Scroll for more
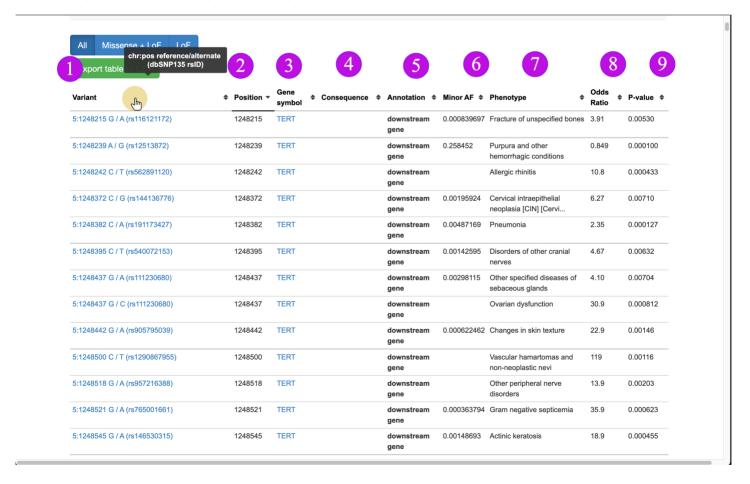Scroll down to reveal more result

8 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 5, position 1,248,215)
   b. Genetic alteration (here: altered from nucleotide G to A)
   c. rsID (here: rs116121172)
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

Variant	Position	Gene symbol	Consequence	Annotation	Minor AF	Phenotype	Odds Ratio	P-value
5:1248215 G / A (rs116121172)	1248215	TERT		downstream gene	0.000839697	Fracture of unspecified bones	3.91	0.00530
5:1248239 A / G (rs12513872)	1248239	TERT		downstream gene	0.258452	Purpura and other hemorrhagic conditions	0.849	0.000100
5:1248242 C / T (rs562891120)	1248242	TERT		downstream gene		Allergic rhinitis	10.8	0.000433
5:1248372 C / G (rs144136776)	1248372	TERT		downstream gene	0.00195924	Cervical intraepithelial neoplasia [CIN] [Cervi...	6.27	0.00710
5:1248382 C / A (rs191173427)	1248382	TERT		downstream gene	0.00487169	Pneumonia	2.35	0.000127
5:1248395 C / T (rs540072153)	1248395	TERT		downstream gene	0.00142595	Disorders of other cranial nerves	4.67	0.00632
5:1248437 G / A (rs111230680)	1248437	TERT		downstream gene	0.00298115	Other specified diseases of sebaceous glands	4.10	0.00704
5:1248437 G / C (rs111230680)	1248437	TERT		downstream gene		Ovarian dysfunction	30.9	0.000812
5:1248442 G / A (rs905795039)	1248442	TERT		downstream gene	0.000622462	Changes in skin texture	22.9	0.00146
5:1248500 C / T (rs1290867955)	1248500	TERT		downstream gene		Vascular hamartomas and non-neoplastic nevi	119	0.00116
5:1248518 G / A (rs957216388)	1248518	TERT		downstream gene		Other peripheral nerve disorders	13.9	0.00203
5:1248521 G / A (rs765001661)	1248521	TERT		downstream gene	0.000363794	Gram negative septicemia	35.9	0.000623
5:1248545 G / A (rs146530315)	1248545	TERT		downstream gene	0.00148693	Actinic keratosis	18.9	0.000455

9 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