

# 1 Search by Variant

## General Instruction

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

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

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## 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/variant/9-97793827-G-A>

## 2 Insert SNP Variant

### General Instruction

Alternatively, you can type the **SNP Variant** in the **Search Box** blank space

### Search Format

rsID: e.g., rs965513



### Search Box Colorado Biobank Portal

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

#### Supported queries

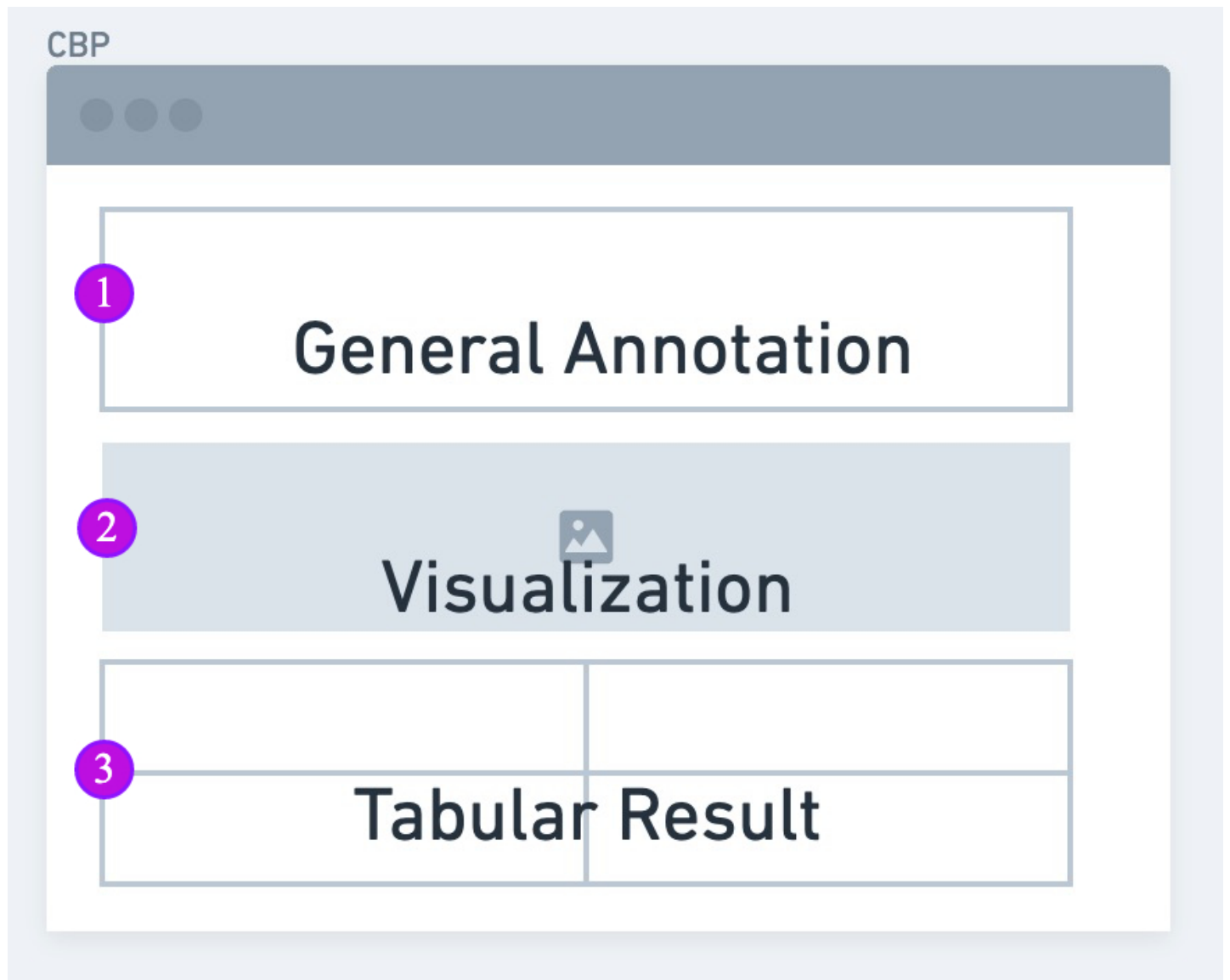
**By gene:** [NRAS](#)  
**By transcript:** [ENST00000310581](#)  
**By variant:** [rs965513](#)  
**By region:** [chr22:17440016-17459999](#)  
**By phenotype:** [Thyroid cancer](#)

## 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.



The diagram illustrates the layout of search results within a window titled "CBP". The window has a dark grey header bar with three white circular icons. Below the header, the content is organized into three distinct segments, each marked with a purple circle containing a white number:

- 1 General Annotation:** A white rectangular box with a thin grey border containing the text "General Annotation".
- 2 Visualization:** A light blue rectangular box with a thin grey border containing a small image icon (a square with a triangle and a circle) and the text "Visualization".
- 3 Tabular Result:** A white rectangular box with a thin grey border containing a table structure. The table has two columns and two rows. The text "Tabular Result" is centered in the bottom-right cell of the table.

# 4 General Annotation

## General Instruction

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

1. Click on each external link to study the SNP variant further

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## Variant: 9:97793827 G / A SNP Variant and External Links

**1** **dbSNP** [rs965513](#)  
**UCSC** [chr9:97793827:A:G](#)  
**gnomAD** [chr9:97793827:A:G](#)  
**ClinVar** [Click to search for variant in Clinvar](#)

**Annotations**  
This variant falls on 8 transcripts in 1 genes:

**intron**

- [PTCSC2](#) Transcripts
- 101928337 - NR\_147055.1

PheWAS Analysis: variant chr9:97793827:A:G

Scroll down

# 5 Variants on Other Gene Transcript

## General Instruction

You can also explore variants in other gene transcripts by selecting among the alternative transcript

## Scroll for more

Scroll down to reveal more result

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## Variant: 9:97793827 G / A

**dbSNP** [rs965513](#)  
**UCSC** [chr9:97793827:A:G](#)  
**gnomAD** [chr9:97793827:A:G](#)  
**ClinVar** [Click to search for variant in Clinvar](#)

### Annotations

This variant falls on 8 transcripts in 1 genes:

**intron**

- PTCSC2
- 10192833

Transcripts

- ENST00000430058
- ENST00000648027
- ENST00000648505
- ENST00000648625
- ENST00000649253
- ENST00000649461 \*
- ENST00000649526
- ENST00000650104

PheWAS Analysis: variant chr9:97793827:A:G

<https://hdc-sandbox-bioengine.uw.r.appspot.com/transcript/ENST00000430058>

## 6 Visualization: PheWAS

### General Instruction

This segment shows the graphical representation of a PheWAS analysis based on the variant of interest

1. The X-axis shows: Negative log<sub>10</sub>(p-value)
2. The Y-axis shows all computed phenotype (listed as phenodes)
3. Mouse over the peak to show the PheWAS summary statistics

