

1 Search by Region

General Instruction

You can search the Colorado Biobank Portal (CBP) using your chromosome region of interest

The easiest way to try this out is to search **By region**

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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/region/22-17440016-17459999>

2 Insert Chromosome Region

General Instruction

Alternatively, you can type the **Chromosome Region** in the **Search Box** blank space

Search Format

Chromosome number:Start position - Stop position

Note

Search region limit: 50,000 base pairs

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Search Box

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3 Visualization

General Instruction

Graphical representation of Chromosome 22 and the SNPs found on this chromosome. The height of the peaks shows $-\log_{10}(p\text{-value})$ of these SNPs.

Scroll for more

Scroll down to reveal more result

Colorado Biobank Portal Gene, transcript, variant tzl.phang@cuaneschutz.edu [Sign out](#) [Terms](#) [Contact](#) [FAQ](#) [Credits](#)

Region: 22 / 17440016 / 17459999

Coverage summary **Graphical Chromosome representation for SNP**

6
5
4
3
2
1
0

17,442,000 17,444,000 17,446,000 17,448,000 17,450,000 17,452,000 17,454,000 17,456,000 17,458,000

Genes
.

Coverage summary

Variants
All Missense + LoF LoF

Scroll down

4 Tabular Summary Statistics

General Instruction

The table shows summary statistics for all the SNPs

1. **Variants:** Single Nucleotide Polymorphism (SNP), labeled with these genetic info:
 - a. Chromosome location (here: Chromosome 22, position 17,440,206)
 - b. Genetic alteration (here: altered from nucleotide A to G)
 - c. rsID (here: rs1350617173)
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
22:17440206 A / G (rs1350617173)	17440206	CECR2		intron		Parasomnia	61.2	0.00634
22:17440207 C / CTG (rs145052413)	17440207	CECR2		intron	0.114507	Osteoporosis	0.826	0.00153
22:17440287 C / T (rs12628022)	17440287	CECR2		intron	0.0249765	Seborrheic keratosis	1.48	0.00209
22:17440322 G / A (rs5746381)	17440322	CECR2		intron	0.115012	Osteoporosis	0.824	0.00133
22:17440390 C / CT (rs921349602)	17440390	CECR2		intron		Other abnormal glucose	7.99	0.0285
22:17440534 C / T (rs181050797)	17440534	CECR2		intron		Nervous system congenital anomalies	24.4	0.0128
22:17440550 CTTA / C (rs373160591)	17440550	CECR2		intron	0.000814621	Atherosclerosis of aorta	16.7	0.00316
22:17440606 T / A (rs12628376)	17440606	CECR2		intron	0.0467307	Joint effusions	0.639	0.00425
22:17440608 T / C (rs556879688)	17440608	CECR2		intron		Personal history of diseases of digestive system	23.0	0.00130
22:17440610 T / C (rs12159248)	17440610	CECR2		intron	0.00605645	Opiates and related narcotics causing adverse e...	1.72	0.00679
22:17440658 G / A (rs79950448)	17440658	CECR2		intron	0.0214105	Nausea and vomiting	0.720	0.000179
22:17440717 C / T (rs16982467)	17440717	CECR2		intron	0.00188422	Effects of other external causes	4.56	0.0000215
22:17440768 C / T (rs9617583)	17440768	CECR2		intron	0.00263462	Encounter for long-term (current) use of antico...	2.53	0.00122
22:17440788 A / C (rs5747137)	17440788	CECR2		intron	0.115014	Osteoporosis	0.824	0.00133
22:17440823 G / A (rs1424887459)	17440823	CECR2		intron		Chronic ulcer of skin	73.1	0.000173
22:17440828 C / G (rs1008287491)	17440828	CECR2		intron		Mechanical complication of unspecified genitour...	164	0.000649
22:17440951 G / A (rs570414881)	17440951	CECR2		intron		Calculus of bile duct	107	0.00225
22:17440954 G / T (rs190287822)	17440954	CECR2		intron	0.00778567	Influenza	3.11	0.00250
22:17441039 C / T (rs67203878)	17441039	CECR2		intron	0.0213385	Seborrheic keratosis	1.55	0.000806