

Webツール入門

大阪大学大学院医学系研究科 遺伝統計学
東京大学大学院医学系研究科 遺伝情報学
理化学研究所生命医科学研究センター システム遺伝学チーム

<http://www.sg.med.osaka-u.ac.jp/index.html>

Webツール入門

- ① **ゲノム・遺伝子情報のWebツール**
- ② **遺伝子変異・SNP情報のWebツール**
- ③ **疾患感受性遺伝子情報・解析結果のWebツール**
- ④ **エピゲノム情報のWebツール**
- ⑤ **創薬情報のWebツール**

(本講義での紹介内容は、2023年7月現在に動作確認できたWebツールになります。)

①-1:UCSC Genome Browser

UNIVERSITY OF CALIFORNIA SANTA CRUZ UCSC Genome Browser Gateway

Home Genomes Genome Browser Tools Mirrors Downloads My Data Help About Us

Browse/Select Species

POPULAR SPECIES

Human Mouse Rat Fruitfly Worm Yeast

Enter species or common name

REPRESENTED SPECIES

Human Chimp Bonobo Gorilla Orangutan Gibbon Crab-eating macaque Rhesus Baboon (anubis) Baboon (hamadryas) Marmoset Squirrel monkey Tarsier Mouse lemur Bushbaby

Find Position

Human Assembly
Dec. 2013 (GRCh38/hg38)

Position/Search Term
Enter position, gene symbol or search terms
Current position: chr9:133,252,000-133,280,861

GO

Human Genome Browser - hg38 assembly

view sequences

UCSC Genome Browser assembly ID: hg38
Sequencing/Assembly provider ID: GRCh38 Genome Reference Consortium Human Reference 38 (GCA_000001405.15)
Assembly date: Dec. 2013
GenBank accession ID: GCA_000001305.2
NCBI Genome information: NCBI genome/51 (Homo sapiens)
NCBI Assembly information: NCBI assembly/883148 (GRCh38/GCA_000001405.15)
BioProject information: NCBI Bioproject: 31257

Homo sapiens
(Graphic courtesy of CBSE)

Search the assembly:

- By position or search term: Use the "position or search term" box to find areas of the genome associated with many different attributes, such as a specific chromosomal coordinate range; mRNA, EST, or STS marker names; or keywords from the GenBank description of an mRNA. **More information**, including sample queries.
- By gene name: Type a gene name into the "search term" box, choose your gene from the drop-down list, then press "submit" to go directly to the assembly location associated with that gene. **More information**.
- By track type: Click the "track search" button to find Genome Browser tracks that match specific selection criteria. **More information**.

<https://genome.ucsc.edu/cgi-bin/hgGateway>

•ゲノム配列の標準的な閲覧サイトです。

•”KLF4”と入力してみましょう。

①-1:UCSC Genome Browser

The screenshot displays the UCSC Genome Browser interface for the human genome (GRCh38/hg38) assembly. The main view is centered on chromosome 9, specifically the region chr9:107,484,852-107,489,646 (4,795 bp). The interface includes a navigation bar at the top with options like Genomes, Genome Browser, Tools, Mirrors, Downloads, My Data, View, Help, and About Us. Below the navigation bar, there are controls for moving and zooming the view, including a search bar for positions, gene symbols, or search terms. The main display area shows multiple tracks of genomic data, including the KLF4 gene structure, RefSeq Genes, Human mRNAs, H3K27Ac marks, DNase I Hypersensitivity Peak Clusters, and Multiz Alignments of 100 Vertebrates. The bottom of the interface features a control panel with buttons for track search, default tracks, default order, hide all, add custom tracks, track hubs, configure, multi-region, reverse, resize, refresh, collapse all, and expand all.

<https://genome.ucsc.edu/cgi-bin/hgGateway>

・KLF4遺伝子周辺のヒトゲノム領域において、塩基配列・遺伝子情報・エピゲノム情報・SNP、等の情報を閲覧することができます。

①-2:NCBI Gene

NCBI Resources How To Sign in to NCBI

Gene Gene Search Advanced Help

Gene

Gene integrates information from a wide range of species. A record may include nomenclature, Reference Sequences (RefSeqs), maps, pathways, variations, phenotypes, and links to genome-, phenotype-, and locus-specific resources worldwide.

Using Gene

- [Gene Quick Start](#)
- [FAQ](#)
- [Download/FTP](#)
- [RefSeq Mailing List](#)
- [Gene News](#)
- [Factsheet](#)

Gene Tools

- [Submit GeneRIFs](#)
- [Submit Correction](#)
- [Statistics](#)
- [BLAST](#)
- [Genome Workbench](#)
- [Splign](#)

Other Resources

- [OMIM](#)
- [RefSeq](#)
- [RefSeqGene](#)
- [Protein Clusters](#)

Representative queries

| Find genes by... | Search text |
|-----------------------|--|
| free text | human muscular dystrophy |
| chromosome and symbol | (1[chr]_OR 2[chr]) AND adh*[sym] |

<http://www.ncbi.nlm.nih.gov/gene/>

- 遺伝子情報を集約した標準的なデータベースです (by 米国NCBI)。
- “KLF4” と入力してみましょう。

①-2: NCBI Gene

NCBI Resources How To Sign in to NCBI

Gene Gene [v] [input] Search Advanced Help

Full Report [v] Send to: [v] Hide sidebar >>

KLF4 Kruppel like factor 4 [*Homo sapiens* (human)]

Gene ID: 9314, updated on 26-Jul-2021

Summary [v] [?] [?]

Official Symbol KLF4 provided by [HGNC](#)

Official Full Name Kruppel like factor 4 provided by [HGNC](#)

Primary source [HGNC:HGNC:6348](#)

See related [Ensembl:ENSG00000136826](#) [MIM:602253](#)

Gene type protein coding

RefSeq status REVIEWED

Organism [Homo sapiens](#)

Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo

Also known as EZF; GKLF

Summary This gene encodes a protein that belongs to the Kruppel family of transcription factors. The encoded zinc finger protein is required for normal development of the barrier function of skin. The encoded protein is thought to control the G1-to-S transition of the cell cycle following DNA damage by mediating the tumor suppressor gene p53. Mice lacking this gene have a normal appearance but lose weight rapidly, and die shortly after birth due to fluid evaporation resulting from compromised epidermal barrier function. Alternative splicing results in multiple transcript variants encoding different isoforms. [provided by RefSeq, Sep 2015]

Expression Broad expression in colon (RPKM 75.2), esophagus (RPKM 73.6) and 22 other tissues [See more](#)

Orthologs [mouse](#) [all](#)

NEW Try the new [Gene table](#)
Try the new [Transcript table](#)

Table of contents [v]

- Summary
- Genomic context
- Genomic regions, transcripts, and products
- Expression
- Bibliography
- Phenotypes
- Variation
- Pathways from PubChem
- Interactions
- General gene information
 - Markers, Related pseudogene(s), Homology, Gene Ontology
- General protein information
- NCBI Reference Sequences (RefSeq)
- Related sequences
- Additional links

Genome Browsers [v]

<http://www.ncbi.nlm.nih.gov/gene/>

• KLF4 遺伝子の基礎的情報や生物学的機能、関連文献、等がリンクと共に提供されます。ゲノム配列ブラウザーも埋め込まれています。

①-3:HGNC (HUGO Gene Nomenclature Committee)

Home Gene data Tools Downloads VGNC Contact us More

Request symbol



The resource for approved human gene nomenclature

Search symbols, keywords or IDs



Last updated: 2021-07-28

The work of the HGNC is supported by National Human Genome Research Institute (NHGRI) grant U24HG003345 & Wellcome Trust grant 208349/Z/17/Z.



Gene data

Gene symbol reports
Gene group reports

Tools

BioMart
HCOP
Multi-symbol checker
Search

About

About the HGNC
Guidelines
HGNC team
Meetings
Privacy notice
Publications
Scientific Advisory Board
Specialist advisors
Workshops

Help

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Help index
Useful links

Downloads

BioMart
Complete set archive
Custom downloads
REST service
Statistics & download files

News

HGNC announcements
Genenames blog
Current newsletter
Newsletter archive

Contact us

Details
Feedback
Request symbol

<http://www.genenames.org/>

• 遺伝子の名称をまとめた公式サイトです。

• "KLF4"と入力してみましょう。

①-3:HGNC (HUGO Gene Nomenclature Committee)

HGNC

Search symbols, keywords or IDs

Home Gene data Tools Downloads VGNC Contact us More

Request symbol

Symbol report for KLF4

Report

HCOP homology predictions

HGNC data for KLF4

Approved symbol [?](#) KLF4
Approved name [?](#) Kruppel like factor 4
Locus type [?](#) gene with protein product
HGNC ID [?](#) HGNC:6348
Symbol status [?](#) Approved
Previous names [?](#) " Kruppel-like factor 4 (gut) "
Alias symbols [?](#) EZF; GKLF
Alias names [?](#) " gut Kruppel-like factor "
Chromosomal location [?](#) 9q31.2
Gene groups [?](#) Kruppel like factors
Zinc fingers C2H2-type

Gene resources for KLF4

| | | | |
|---------|---|------------------------------|--|
| Ensembl | ENSG00000136826 Curated | NCBI Gene | 9314 Curated |
| | Ensembl region in detail | | |
| | Ensembl gene sequence | | |
| UCSC | uc004bdg.4 | Alliance of Genome Resources | HGNC:6348 |

<http://www.genenames.org/>

・KLF4遺伝子が、正式名称”KLF4”の他に、”EZF”、”GKLF”、”gut Kruppel-like factor”などの名称で呼ばれていたことがわかります。⁸

①-4:EMBL-EBI

EMBL-EBI

Services

Research

Training

About us

EMBL-EBI

The EMBL-EBI website has been redesigned. Please send us feedback about this page.

EMBL's European Bioinformatics Institute

EMBL-EBI

Unleashing the potential of big data in biology



Search

Example searches: blast keratin bfl1 | About EBI Search

Find data resources →

Submit data →

Explore our research →

Train with us →

Latest news →

<http://www.ebi.ac.uk/>

• 遺伝子情報を集約した標準的なデータベースです (by 欧州EBI)。

• "KLF4" と入力してみましょう。

①-4:EMBL-EBI

KLF4 expression summary

View in Expression Atlas [→](#)

Baseline expression

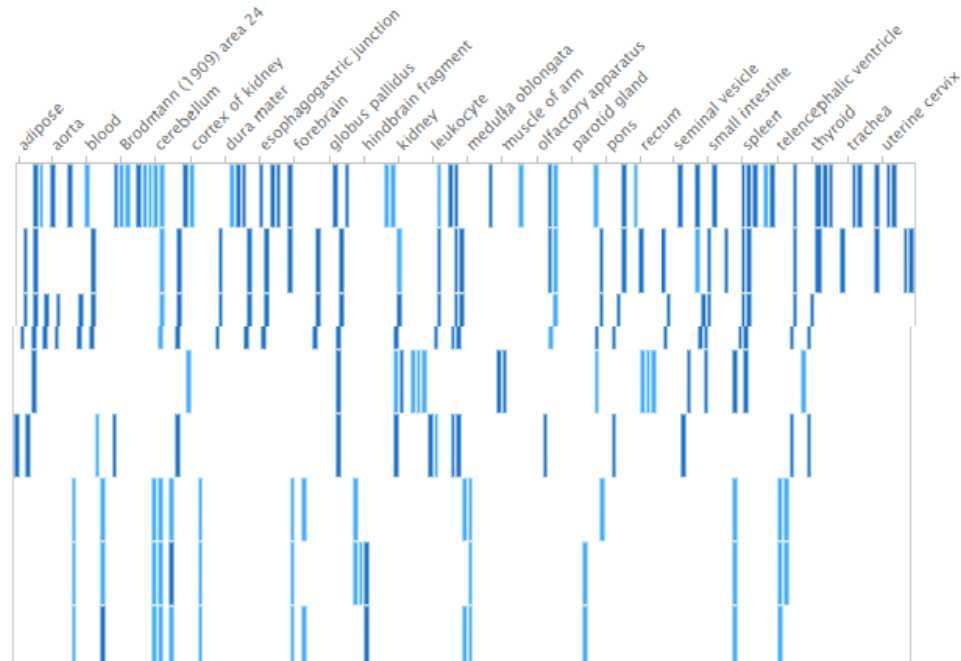
Differential expression

Showing 35 experiments:

By experiment type ▾ Filters Download



- [GTEx](#)
- [32 Uhlen's Lab](#)
- [Hallstrom et al., 2014 - Organism part](#)
- [19 NIH Epigenomics Roadmap](#)
- [Illumina Body Map](#)
- [HDBR developing brain - 9 post ...](#)
- [HDBR developing brain - 12 post ...](#)
- [HDBR developing brain - Carnegie Stage ...](#)



<http://www.ebi.ac.uk/>

•KLF4遺伝子の基礎的情報や生物学的機能、発現情報、蛋白質情報、関連文献、等がリンクと共に提供されます。

①-5:Ensembl

Tools [BioMart >](#) [BLAST/BLAT >](#) [Variant Effect Predictor >](#)

[All tools](#) Export custom datasets from Ensembl with this data-mining tool

Search our genomes for your DNA or protein sequence

Analyse your own variants and predict the functional consequences of known and unknown variants

Ensembl is a genome browser for vertebrate genomes that supports research in comparative genomics, evolution, sequence variation and transcriptional regulation. Ensembl annotate genes, computes multiple alignments, predicts regulatory function and collects disease data. Ensembl tools include BLAST, BLAT, BioMart and the Variant Effect Predictor (VEP) for all supported species.

Ensembl Release 104 (May 2021)

- Update to the Ensembl Canonical transcript set.
- Human and mouse gene sets updated to GENCODE 38 and GENCODE M27, respectively.
- Retirement of gene names derived from BAC clones.

[More release news](#) on our blog

<http://asia.ensembl.org/index.html>

- 遺伝子情報を集約した標準的なデータベースです (by 欧州Ensembl)。
- “KLF4” と入力してみましょう。

①-5: Ensembl

The screenshot displays the Ensembl genome browser interface for the KLF4 gene. At the top, the Ensembl logo is visible along with navigation links for BLAST/BLAT, BioMart, Tools, Downloads, Help & Documentation, Blog, and Mirrors. A search bar is located in the top right corner. The main navigation bar shows the current context: Human (GRCh38.p7), Location: 9:107,484,852-107,490,482, and Gene: KLF4.

The left sidebar contains a tree view of 'Gene-based displays' with categories such as Summary, Sequence, Comparative Genomics, Ontologies, Phenotypes, Genetic Variation, Regulation, External references, Supporting evidence, and ID History.

The main content area is titled 'Gene: KLF4 ENSG00000136826' and includes the following sections:

- Description:** Kruppel like factor 4 [Source:HGNC Symbol;Acc:HGNC:6348]
- Synonyms:** GKLF, EZF
- Location:** Chromosome 9: 107,484,852-107,490,482 reverse strand. GRCh38:CM000671.2
- About this gene:** This gene has 6 transcripts (splice variants), 57 orthologues, 11 paralogues, is a member of 1 Ensembl protein family and is associated with 14 phenotypes.
- Transcripts:** A button labeled 'Show transcript table' is present.
- Summary:** A section with a question mark icon.
- Name:** KLF4 (HGNC Symbol)
- CCDS:** This gene is a member of the Human CCDS set: CCDS6770.2
- UniProtKB:** This gene has proteins that correspond to the following UniProtKB identifiers: O43474
- RefSeq:** Overlapping RefSeq annotation not matched
- Ensembl version:** ENSG00000136826.14
- Other assemblies:** This gene maps to 110,247,133-110,252,763 in GRCh37 coordinates. View this locus in the GRCh37 archive: ENSG00000136826
- Gene type:** Known protein coding
- Annotation method:** Annotation for this gene includes both automatic annotation from Ensembl and Havana manual curation, see article.
- Alternative genes:** This gene corresponds to the following database identifiers: Havana gene: OTTHUMG00000020449

At the bottom of the main content area, there is a button labeled 'Go to Region in Detail for more tracks and navigation options (e.g. zooming)'.

<http://asia.ensembl.org/index.html>

- 他の遺伝子データベース同様、KLF4遺伝子に関連する情報がリンクと共に提供されます。ヒト以外の種のゲノム情報も充実しています。

①-6: miRBase

miRBase: the microRNA database

miRBase provides the following services:

- The [miRBase database](#) is a searchable database of published miRNA sequences and annotation. Each entry in the miRBase Sequence database represents a predicted hairpin portion of a miRNA transcript (termed mir in the database), with information on the location and sequence of the mature miRNA sequence (termed miR). Both hairpin and mature sequences are available for [searching](#) and [browsing](#), and entries can also be retrieved by name, keyword, references and annotation. All sequence and annotation data are also [available for download](#).
- The [miRBase Registry](#) provides miRNA gene hunters with unique names for novel miRNA genes prior to publication of results. Visit the [help pages](#) for more information about the naming service.

To receive email notification of data updates and feature changes please subscribe to the [miRBase announcements mailing list](#). Any queries about the website or naming service should be directed at mirbase@manchester.ac.uk.

miRBase is managed by the [Griffiths-Jones lab](#) at the [Faculty of Biology, Medicine and Health, University of Manchester](#) with funding from the [BBSRC](#). miRBase was previously hosted and supported by the [Wellcome Trust Sanger Institute](#).

References

If you make use of the data presented here, please cite the following articles in addition to the primary data sources:

[miRBase: from microRNA sequences to function.](#)
Kozomara A, Birgaoanu M, Griffiths-Jones S.
Nucleic Acids Res 2019 47:D155-D162

[miRBase: annotating high confidence microRNAs using deep sequencing data.](#)
Kozomara A, Griffiths-Jones S.
Nucleic Acids Res 2014 42:D68-D73

[miRBase: integrating microRNA annotation and deep-sequencing data.](#)
Kozomara A, Griffiths-Jones S.
Nucleic Acids Res 2014 42:D152-D157

<http://www.mirbase.org/>

• マイクロRNA情報を集約した標準的なデータベースです。

• ”[hsa-mir-146a](#)”と入力してみましょう。

①-6: miRBase

miRBase MANCHESTER 1824

Home Search Browse Help Download Blog Submit **hsa-mir-146a** submit

Stem-loop sequence hsa-mir-146a


Accession MI0000477 ([change log](#))

Previous IDs hsa-mir-146

Symbol [HGNC:MIR146A](#)

Description *Homo sapiens* miR-146a stem-loop

Gene family MIPF0000103; [mir-146](#)


Literature search  [749 open access papers](#) mention hsa-mir-146a (6672 sentences)

Stem-loop

```
5'   c   -----u   u   uu   c   u   g   uc
    cgaug   guaucc   cagcu   gagaacugaauu   ca   ggguu   ug   a
    |||||   |||||   |||||   |||||   |||||   |||||   |||||   |||||   |||||   |||||
3'   gcuac   uauagg   gucga   uucugacuuaa   gu   uccag   ac   u
    u   ugucuc   -   -c   a   c   -   ug
```

[Get sequence](#)

Deep sequencing [1355616](#) reads, [2.37e+03](#) reads per million, 160 experiments



Confidence Annotation confidence: high

<http://www.mirbase.org/>

・マイクロRNA mir146aの基礎的情報や各種ID、標的遺伝子情報、疾患との関連、生物学的機能、関連文献、等がリンクと共に提供されます。

①-7:IMGT/HLA

IPD-IMGT/HLA

Overview **IMGT/HLA** KIR MHC NHKIR HPA ESTDAB

IPD / IMGT/HLA

Welcome to IPD-IMGT/HLA

Release 3.49 (2022-07) Version Report - Build 536e883

The IPD-IMGT/HLA Database provides a specialist database for sequences of the human major histocompatibility complex (MHC) and includes the official sequences named by the [WHO Nomenclature Committee For Factors of the HLA System](#). The IPD-IMGT/HLA Database was originally part of the international ImMunoGeneTics project (IMGT). For more information about the database and what data and tools are available please see our [about](#) page.

Alignment

The alignment tool provides access to pre-compiled alignments for individual HLA genes and sequence features >

Alleles

Query the IPD-IMGT/HLA database for officially named alleles with the allele query tool. >

Statistics

Latest IPD-IMGT/HLA Statistics and Release reports >

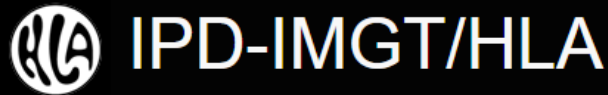
Download

Access to the IPD-IMGT/HLA ftp for downloading sequence files >

<http://www.ebi.ac.uk/ipd/imgt/hla/>

- HLA遺伝子配列情報(=白血球の血液型)を集約したデータベースです。
- "Alleles"のページで、"A*01"と入力してみましょう。

①-7:IMGT/HLA



Overview **IMGT/HLA** KIR MHC NHKIR HPA ESTDAB

IPD / IMGT/HLA / ALLELE LIST / ALLELE

Allele Report for A*01:01:01:01

View

Primary Name: A*01:01:01:01

| | | | |
|------------------------|--|-----------------|---------------------|
| Pre-2010 Nomenclature: | A*01010101 | Aliases: | A*01011, A*0101, A1 |
| IMGT/HLA Acc No: | HLA00001 | OMIM Entry: | 142800 |
| Assigned: | 1989-08-01 | Last updated: | 1998-12-16 |
| Allele G Group: | A*01:01:01G | Allele P Group: | A*01:01P |
| INSDC Source Entries | AJ278305 AL645935 CR759913 EU445470 GU812295 HG794373 M24043 X55710 Z93949 | | |
| | COX | Ethnicity: | Caucasoid |
| | MOLT-4 | Ethnicity: | Unknown |
| | 7550800303 | Ethnicity: | Oriental |

<http://www.ebi.ac.uk/ipd/imgt/hla/>

•HLA-A遺伝子の遺伝子配列の一つ、A*01:01:01:01について、塩基配列やアミノ酸配列の情報が得られます。

Webツール入門

- ① ゲノム・遺伝子情報のWebツール
- ② 遺伝子変異・SNP情報のWebツール
- ③ 疾患感受性遺伝子情報・解析結果のWebツール
- ④ エピゲノム情報のWebツール
- ⑤ 創薬情報のWebツール

②-1 : dbSNP

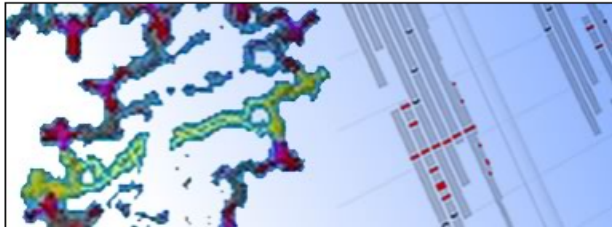
dbSNP

SNP

Advanced

Search

Help



dbSNP

dbSNP contains human single nucleotide variations, microsatellites, and small-scale insertions and deletions along with publication, population frequency, molecular consequence, and genomic and RefSeq mapping information for both common variations and clinical mutations.

Getting Started

[dbSNP 20th Anniversary](#)

[Overview of dbSNP](#)

[About Reference SNP \(rs\)](#)

[Factsheet](#)

[Entrez Updates \(May 26, 2020\)](#)

Submission

[How to Submit](#)

[Hold Until Published \(HUP\) Policies](#)

[Submission Search](#)

Access Data

[Web Search](#)

[eUtils API](#)

[Variation Services](#)

[FTP Download](#)

[Tutorials on GitHub](#)

<http://www.ncbi.nlm.nih.gov/SNP/>

- SNP情報を集約した標準的なデータベースです (by 米国NCBI)。
- ”rs671” と入力してみましょう。

②-1 : dbSNP

Reference SNP (rs) Report

[Download](#) [f](#) [t](#) [g+](#) [?](#)

[← Switch to classic site](#)

rs671

Current Build 155
Released April 9, 2021

| | | | |
|-----------------------|--|------------------------------|---|
| Organism | <i>Homo sapiens</i> | Clinical Significance | Reported in ClinVar |
| Position | chr12:111803962 (GRCh38.p13) ? | Gene : Consequence | ALDH2 : Missense Variant |
| Alleles | G>A | Publications | 245 citations LitVar 603 |
| Variation Type | SNV Single Nucleotide Variation | Genomic View | See rs on genome |
| Frequency | A=0.006055 (1921/317236, ALFA) A=0.008958 (2371/264690, TOPMED) A=0.018882 (4582/242666, GnomAD_exome) (+ 15 more) | | |

Variant Details

Genomic Placements [?](#)

Clinical Significance

Frequency

HGVS

Submissions

Sequence name

Change

ALDH2 RefSeqGene

NG_012250.2:g.42076G>A

GRCh37.p13 chr 12

NC_000012.11:g.112241766G>A

GRCh38.p13 chr 12

NC_000012.12:g.111803962G>A

<http://www.ncbi.nlm.nih.gov/SNP/>

•ALDH2遺伝子上のアミノ酸置換を伴うSNP:rs671について、変異情報や、遺伝子上の位置、各人類集団での頻度などの情報が提供されます。¹⁹

②-2: ClinVar

NCBI Resources How To Sign in to NCBI

ClinVar ClinVar Search ClinVar for gene symbols, HGVS expressions, conditions, and more Search

Advanced Help

Home About Access Help Submit Statistics FTP

ACTGATGGTATGGGGCCAAGAGATATATCT
CAGGTACGGCTGTCATCACTTAGACCTCAC
CAGGGCTGGGCATAAAAGTCAGGGCAGAGC
CCATGGTGCATCTGACTCCTGAGGAGAAGT
GCAGGTTGGTATCAAGGTTACAAGACAGGT
GGCACTGACTCTCTGCCTATTGGTCTAT

ClinVar

ClinVar aggregates information about genomic variation and its relationship to human health.

Using ClinVar

- [About ClinVar](#)
- [Data Dictionary](#)
- [Downloads/FTP site](#)
- [FAQ](#)
- [Contact Us](#)
- [Factsheet](#)

Tools

- [ACMG Recommendations for Reporting of Incidental Findings](#)
- [ClinVar Submission Portal](#)
- [Submissions](#)
- [Variation Viewer](#)
- [Clinical Remapping - Between assemblies and RefSeqGenes](#)
- [RefSeqGene/LRG](#)

Related Sites

- [ClinGen](#)
- [GeneReviews @](#)
- [GTR @](#)
- [MedGen](#)
- [OMIM @](#)
- [Variation](#)

Submitter highlights

We gratefully acknowledge those who have submitted data and provided advice during the development of ClinVar.

Follow us on [Twitter](#) to receive announcements of the release of new datasets.

Want to learn more about who submits to ClinVar?

<http://www.ncbi.nlm.nih.gov/clinvar/>

- 疾患リスクに重点を置いてSNP情報を集約したデータベースです。
- ”rs671”と入力してみましよう。

②-2: ClinVar

NM_000690.4(ALDH2):c.1510G>A (p.Glu504Lys)

Cite this record

Interpretation: drug response

Review status: ★★☆☆ reviewed by expert panel

Submissions: 7

First in ClinVar: Apr 4, 2013

Most recent Submission: Jul 8, 2022

Last evaluated: Mar 24, 2021

Accession: VCV000018390.6

Variation ID: 18390

Description: single nucleotide variant

Variant details

Conditions

Gene(s)

NM_000690.4(ALDH2):c.1510G>A (p.Glu504Lys)

Allele ID: 33429

Variant type: single nucleotide variant

Variant length: 1 bp

Chromosomal location: 10q24.12

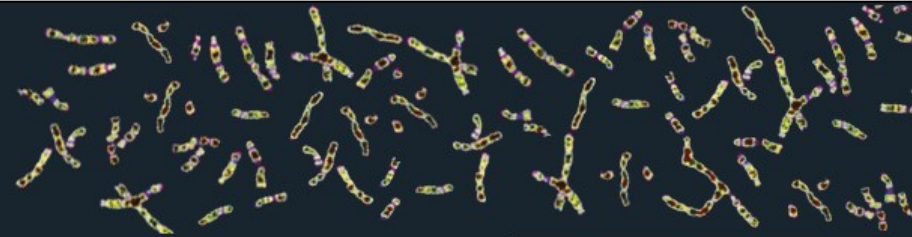
<http://www.ncbi.nlm.nih.gov/clinvar/>

- ALDH2遺伝子上のアミノ酸置換を伴うSNP:rs671について、疾患リスクに重点を置いて情報が提供されます。

②-3: 1000 Genomes Project

IGSR: The International Genome Sample Resource

Supporting open human variation data



Home About Data Help

Search IGSR



The International Genome Sample Resource

The 1000 Genomes Project created a catalogue of common human genetic variation, using openly consented samples from people who declared themselves to be healthy. The reference data resources generated by the project remain heavily used by the biomedical science community.

The International Genome Sample Resource (IGSR) maintains and shares the human genetic variation resources built by the 1000 Genomes Project. We also update the resources to the current reference assembly, add new data sets generated from the 1000 Genomes Project samples and add data from projects working with other openly consented samples.



| Population | A | G | AIA | AIG | GIG |
|------------|-------------|-------------|------------|-------------|-------------|
| ESN | 0.066 (13) | 0.770 (94) | 0.010 (1) | 0.111 (11) | 0.807 (37) |
| GWD | 0.066 (15) | 0.934 (185) | 0.009 (1) | 0.115 (13) | 0.879 (87) |
| LWK | 0.111 (20) | 0.934 (211) | 0.009 (1) | 0.182 (18) | 0.876 (99) |
| MSL | 0.024 (4) | 0.976 (166) | 0.047 (4) | 0.953 (81) | 0.047 (4) |
| YRI | 0.079 (17) | 0.921 (199) | 0.157 (17) | 0.843 (91) | 0.157 (17) |
| AMR | 0.365 (253) | 0.635 (441) | 0.147 (51) | 0.435 (151) | 0.418 (145) |

View variants in genomic context in Ensembl

Latest Announcements

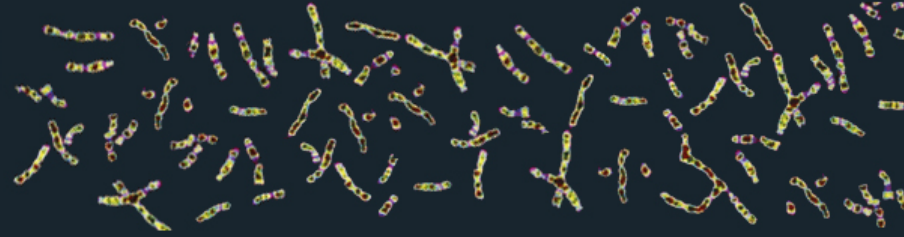
<http://www.internationalgenome.org/>

- 多数の人類集団**2500人**の全ゲノムシーケンス結果を公開しています。
- **“Data”**のページに飛んでみましょう。

②-3: 1000 Genomes Project

IGSR: The International Genome Sample Resource

Supporting open human variation data



[Home](#) [About](#) [Data](#) [Help](#)

Search IGSR



Using data from IGSR

IGSR provides open data to support the community's research efforts. You can see our terms of use in our [data disclaimer](#). Please also consult the associated data reuse statements and cite associated publications appropriately. To cite IGSR, please use our [NAR paper](#).

Explore the data sets in IGSR through our [data portal](#)

IGSR shares data files from many studies via our FTP site. To make it easier to find the files you want, we present key data sets in our [data portal](#).

Files can be browsed by:

- sample (i.e. NA12878)
- population (i.e. Yoruba in Ibadan, Nigeria)
- technology (i.e. PacBio HiFi)
- data type (i.e. alignment)
- collection (i.e. 1000 Genomes Project phase three)

Our portal provides an [overview of the available collections and their associated publications](#).

View variants in genomic context in [Ensembl](#)

IGSR works alongside the [Ensembl genome browser](#). Ensembl presents some of the key call sets in IGSR, placing the variation data in genomic context and adding up-to-date annotation of the

<http://www.internationalgenome.org/>

• 全ゲノムシークエンスの結果得られた遺伝子変異のデータベースを、直接ダウンロードすることができます。

②-4: gnomAD Browser

gnomAD browser

gnomAD v2.1.1

Search

[About](#) [News](#) [Downloads](#) [Terms](#) [Publications](#) [Feedback](#) [Changelog](#) [Help](#)

We're hiring! • Computational scientist



Genome Aggregation Database

gnomAD v2.1.1

Search by gene, region, or variant

Or

- [Find co-occurrence of two variants](#)
- [Download gnomAD data](#)
- [Read gnomAD publications](#)

Please note that gnomAD v2.1.1 and v3.1.2 have substantially different but overlapping sample compositions and are on different genome builds. For more information, see "[Should I switch to the latest version of gnomAD?](#)"

Examples

<https://gnomad.broadinstitute.org/>

- 複数集団 **13万人** の全エクソームシーケンス結果を公開しています。
- **"ALDH2"** と入力してみましょう。

②-4: gnomAD Browser

gnomAD browser

gnomAD v2.1.1

Search

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We're hiring! • Computational scientist

ALDH2 aldehyde dehydrogenase 2 family member

Dataset gnomAD v2.1.1 gnomAD SVs v2.1

Genome build GRCh37 / hg19

Ensembl gene ID ENSG00000111275.8

Ensembl canonical transcript ENST00000261733.2

Other transcripts ENST00000549106.1, ENST00000548536.1, ENST00000416293.3

Region 12:112204691-112247782

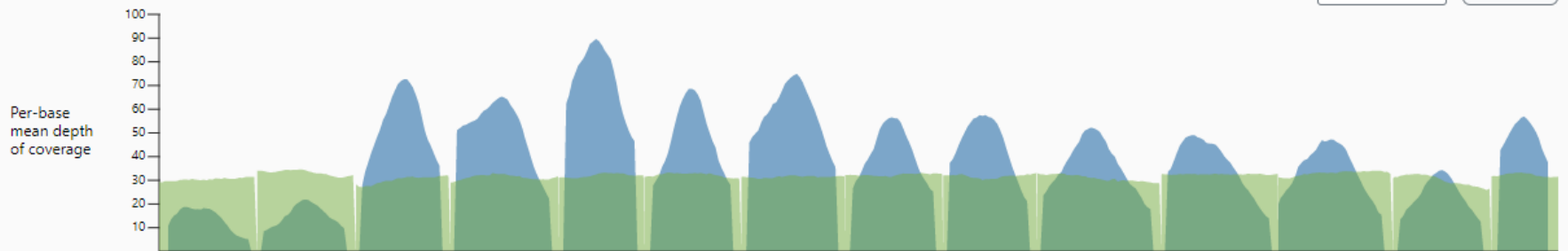
External resources Ensembl, UCSC Browser, and more

Constraint

| Category | Expected SNVs | Observed SNVs | Constraint metrics |
|------------|---------------|---------------|---------------------------------------|
| Synonymous | 134.9 | 143 | Z = -0.55 o/e = 1.06 (0.92 - 1.22) |
| Missense | 316.1 | 250 | Z = 1.32 o/e = 0.79 (0.71 - 0.88) |
| pLoF | 24.4 | 18 | pLI = 0 o/e = 0.74 (0.51 - 1.09) |

Constraint metrics based on Ensembl canonical transcript (ENST00000261733.2).

Viewing full gene. Zoom in



<https://gnomad.broadinstitute.org/>

• ALDH2遺伝子上のSNPについて、欧米人集団およびアフリカ系集団における頻度情報が提供されます。

②-5: MGeND (Medical Genomics Japan Variant Database)



MGeND

Variants ▾ Download Help News About ▾

MGeND

Medical Genomics Japan Variant Database

free word

Example - BRAF, E542K, Lynch syndrome

Select category :

ANNOTATION

Variant annotation service.

<https://mgend.ncgm.go.jp/>

- 日本人集団における機能性遺伝子変異をまとめたデータベースです。
- “BRAF”と入力してみましょう。

②-5: MGeND (Medical Genomics Japan Variant Database)

| Variant name | Gene | AA change | MGeND | | | | ClinVar |
|--|------|-----------|-------|---------|---------|------------------------|---|
| | | | Entry | Origin | Type | Annotation | |
| NC_000007.13:g.14048137_6_140481377insT | BRAF | | 1 | unknown | Variant | not provided | |
| NC_000007.14:g.14075334_5A>C | BRAF | p.L597R | 1 | somatic | Variant | Pathogenic | Pathogenic Likely pathogenic ★★★★ |
| NC_000007.13:g.14048141_1C>G (rs121913351) | BRAF | p.G466A | 1 | somatic | Variant | Pathogenic | Likely pathogenic ★★★★ |
| NC_000007.13:g.14048141_1C>T (rs121913351) | BRAF | p.G466E | 1 | somatic | Variant | Pathogenic | Likely pathogenic ★★★★ |
| NC_000007.14:g.14078160_2C>T | BRAF | p.G469E | 1 | somatic | Variant | Pathogenic | Pathogenic Likely pathogenic ★★★★ |
| NC_000007.13:g.14048736_0G>A (rs397507472) | BRAF | p.R389C | 1 | somatic | Variant | Uncertain significance | Uncertain significance ★★★★ |
| NC_000007.13:g.14053459_7C>T (rs749247588) | BRAF | p.G106R | 1 | somatic | Variant | not provided | Uncertain significance ★★★★ |
| NC_000007.13:g.14045313_6_140453137delinsCT (rs121913227) | BRAF | p.V600R | 1 | somatic | Variant | Pathogenic | Pathogenic ★★★★ |
| NC_000007.13:g.14045317_4G>T (rs121913336) | BRAF | p.D587E | 1 | unknown | Variant | Likely pathogenic | Likely pathogenic ★★★★ |
| NC_000007.13:g.14048137 | BRAF | p.G478S | 1 | unknown | Variant | not provided | |

<https://mgend.ncgm.go.jp/>

- BRAF遺伝子の遺伝子変異リストが、変異の種類や遺伝子機能に与える影響の注釈(annotation)と共に提供されます。

②-6: deCAF

HOME ABOUT TERMS SEND FEEDBACK

deCAF
deCODE Allele Frequency Browser

Search for a gene, variant or region...

Examples:

[PCSK9](#)
[chr13-32398489-A-T](#)
[chr1:55039479-55039500](#)

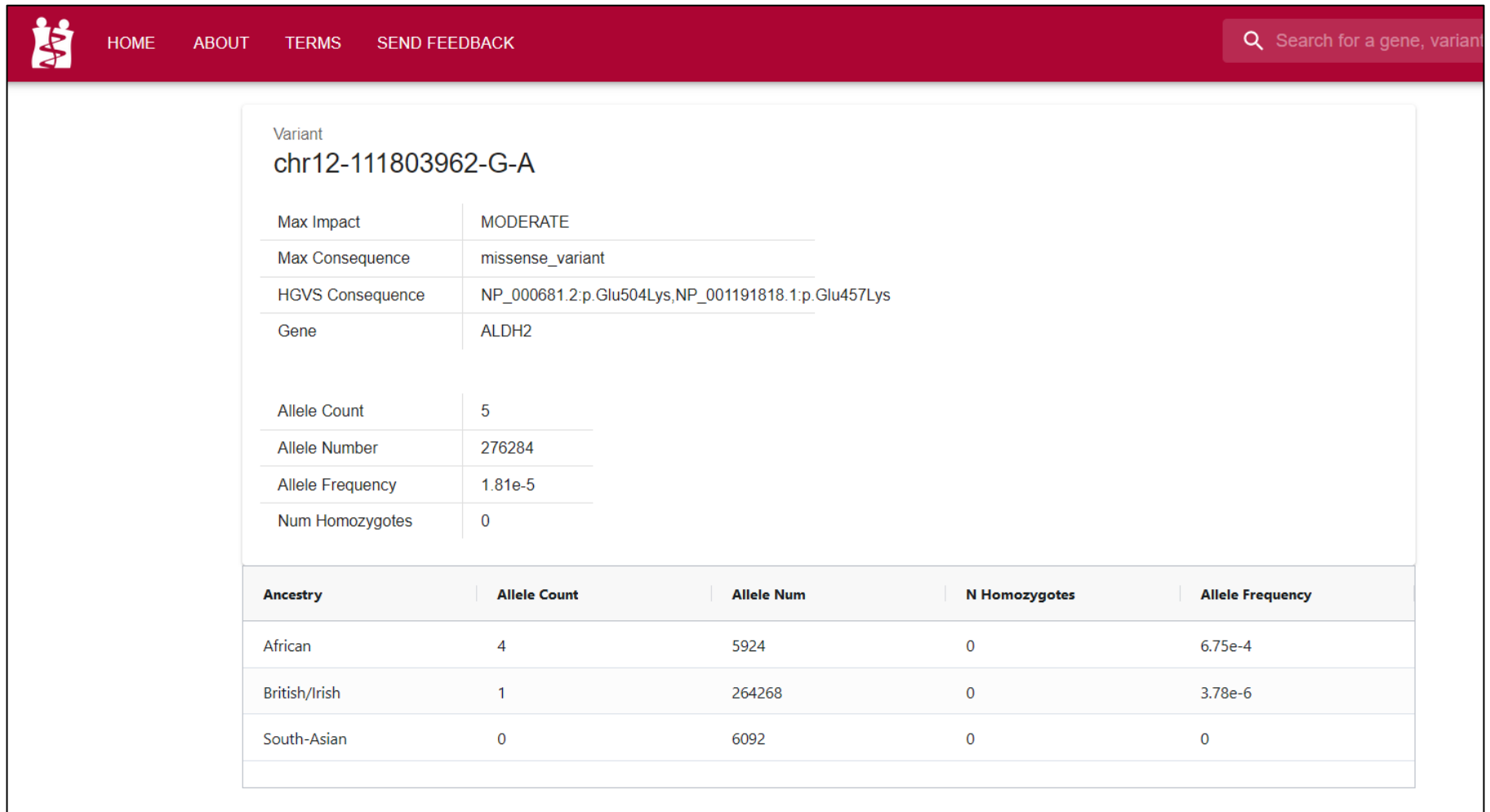
deCAF is a resource of variant allele frequencies made available to the public.
The dataset encompasses SNP and indel variant calls in 150,119 individuals from whole genome sequencing of the UK biobank.

© deCODE genetics

<https://decaf.decode.com/>

- UKバイオバンク15万人の全ゲノムシーケンス結果を公開しています。
- "ALDH2" → "rs671"と入力してみましょう。

②-6: deCAF



The screenshot shows the deCAF website interface. At the top, there is a navigation bar with links for HOME, ABOUT, TERMS, and SEND FEEDBACK. A search bar on the right contains the text "Search for a gene, variant". The main content area displays the variant "chr12-111803962-G-A" and a table of its properties. Below this, there is a table showing the allele frequency across different ancestries.

| Property | Value |
|------------------|--|
| Variant | chr12-111803962-G-A |
| Max Impact | MODERATE |
| Max Consequence | missense_variant |
| HGVS Consequence | NP_000681.2:p.Glu504Lys,NP_001191818.1:p.Glu457Lys |
| Gene | ALDH2 |
| Allele Count | 5 |
| Allele Number | 276284 |
| Allele Frequency | 1.81e-5 |
| Num Homozygotes | 0 |

| Ancestry | Allele Count | Allele Num | N Homozygotes | Allele Frequency |
|---------------|--------------|------------|---------------|------------------|
| African | 4 | 5924 | 0 | 6.75e-4 |
| British/Irish | 1 | 264268 | 0 | 3.78e-6 |
| South-Asian | 0 | 6092 | 0 | 0 |

<https://decaf.decode.com/>

•ALDH2遺伝子上のSNPについて、欧米人集団およびアフリカ系集団における頻度情報が提供されます。

②-7:HaploReg

HaploReg v4.1



HaploReg is a tool for exploring annotations of the noncoding genome at variants on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci. Using LD information from the 1000 Genomes Project, linked SNPs and small indels can be visualized along with chromatin state and protein binding annotation from the Roadmap Epigenomics and ENCODE projects, sequence conservation across mammals, the effect of SNPs on regulatory motifs, and the effect of SNPs on expression from eQTL studies. HaploReg is designed for researchers developing mechanistic hypotheses of the impact of non-coding variants on clinical phenotypes and normal variation.

Update 2015.11.05: Version 4.1 GWAS and eQTL have been updated; a simpler pruning strategy is applied when combining GWAS; and links out to other NHGRI/EBI GWAS hits and GRASP QTL hits are provided.

Update 2015.09.15: Version 4.0 now includes many recent eQTL results including the GTEx pilot, four different options for defining enhancers using Roadmap Epigenomics data, and a complete set of source files for download and local analysis. Older versions available: [v3](#), [v2](#), [v1](#).

[Build Query](#) [Set Options](#) [Documentation](#)

Use one of the three methods below to enter a set of variants. If an r^2 threshold is specified (see the Set Options tab), results for each variant will be shown in a separate table along with other variants in LD. If r^2 is set to NA, only queried variants will be shown, together in one table.

Query
(comma-delimited
list of
rsIDs OR
a single
region as
chrN:start-
end):

or, upload
a text file
(one
refSNP ID
per line):

 参照...

or, select
a GWAS:

クエリ送信

<http://www.broadinstitute.org/mammals/haploreg/haploreg.php>

• SNP同士の連鎖不平衡関係(集団内分布の非独立性)やエピゲノム情報を提供するデータベースです。**rs671**と入力してみましょう。

②-7:HaploReg

delimited list of rsIDs OR a single region as chrN.start-end):
or, upload a text file (one refSNP ID per line):
or, select a GWAS:

Query SNP: **rs671** and variants with $r^2 \geq 0.2$

| chr | pos (hg38) | LD (r ²) | LD (D') | variant | Ref | Alt | AFR freq | AMR freq | ASN freq | EUR freq | SiPhy cons | Promoter histone marks | Enhancer histone marks | DNase | Proteins bound | Motifs changed | NHGRI/EBI GWAS hits | GRASP QTL hits | Selected eQTL hits | GENCODE genes | dbSNP func annot |
|-----|------------|----------------------|---------|-----------------------------|---------|-----|----------|----------|----------|----------|------------|------------------------|------------------------|-----------|----------------|-------------------|---------------------|----------------|-------------------------|---------------|------------------|
| 12 | 111557048 | 0.21 | 0.86 | rs11065950 | A | C | 0.01 | 0.12 | 0.49 | 0.00 | | 4 tissues | 11 tissues | VAS | | Arid5b,Pax-4 | | | | ATXN2 | intronic |
| 12 | 111620462 | 0.21 | 0.87 | rs149673355 | 9-mer A | A | 0.21 | 0.13 | 0.50 | 0.00 | | | | | | 4 altered motifs | | 4 hits | 20kb 3' of RP11-686G8.2 | | |
| 12 | 111647692 | 0.28 | 0.82 | rs11065992 | T | C | 0.00 | 0.01 | 0.40 | 0.00 | | | PLCNT | PLCNT | | p300 | | | BRAP | intronic | |
| 12 | 111672685 | 0.66 | 0.81 | rs3782886 | T | C | 0.00 | 0.00 | 0.22 | 0.00 | | | PANC | BLD | | CEBPB,Nkx2 | 2 hits | | BRAP | synonymous | |
| 12 | 111681367 | 0.66 | 0.83 | rs11066001 | T | C | 0.00 | 0.00 | 0.21 | 0.00 | | | | | | 9 altered motifs | | | BRAP | intronic | |
| 12 | 111702885 | 0.49 | 0.85 | rs11066008 | A | G | 0.00 | 0.00 | 0.29 | 0.00 | | | | SKIN | | AP-1,ERalpha-a | | | ACAD10 | intronic | |
| 12 | 111730205 | 0.88 | 0.94 | rs11066015 | G | A | 0.00 | 0.00 | 0.22 | 0.00 | | | BLD | | | 5 altered motifs | 2 hits | | ACAD10 | intronic | |
| 12 | 111792215 | 1 | 1 | rs4646776 | G | C | 0.00 | 0.00 | 0.22 | 0.00 | | | 13 tissues | ADRL,OVRY | | 6 altered motifs | | | ALDH2 | intronic | |
| 12 | 111803084 | 0.2 | 1 | rs7397491 | G | A | 0.11 | 0.17 | 0.58 | 0.01 | | | | MUS | | GR,LUN-1,SP1 | | | ALDH2 | intronic | |
| 12 | 111803962 | 1 | 1 | rs671 | G | A | 0.00 | 0.00 | 0.22 | 0.00 | | | GI | | | 4 altered motifs | 12 hits | | ALDH2 | missense | |
| 12 | 111806025 | 0.2 | 1 | rs11066026 | A | C | 0.11 | 0.17 | 0.58 | 0.00 | | | | | | 11 altered motifs | | | ALDH2 | intronic | |
| 12 | 111811336 | 0.2 | 1 | rs112605264 | A | C | 0.11 | 0.17 | 0.58 | 0.00 | | | | | | Irf | | | 1.4kb 3' of ALDH2 | | |
| 12 | 111814526 | 0.2 | 1 | rs11066031 | C | T | 0.04 | 0.15 | 0.58 | 0.00 | | | | | | Mrg1::Hoxa9 | | | 1.1kb 3' of RP3-462E2.3 | | |
| 12 | 111842427 | 0.2 | 0.98 | rs4767067 | C | T | 0.07 | 0.15 | 0.57 | 0.01 | | 24 tissues | | | 36 tissues | EBF1 | 6 altered motifs | | | MAPKAPK5-AS1 | |
| 12 | 111887962 | 0.26 | 0.97 | rs16941717 | A | C | 0.00 | 0.01 | 0.50 | 0.00 | | | 5 tissues | SKIN,GI | | Crx | | | MAPKAPK5 | intronic | |
| 12 | 111900120 | 0.8 | 0.9 | rs78069066 | G | A | 0.00 | 0.00 | 0.22 | 0.00 | | | | MUS,BLD | POL2 | | | | 3.6kb 3' of MAPKAPK5 | | |

<http://www.broadinstitute.org/mammals/haploreg/haploreg.php>

- SNP:rs671と周辺のSNPの連鎖不平衡関係や、各SNPの位置がゲノム配列上で、どんなエピゲノム修飾と重なっているかがわかります。

Webツール入門

- ① ゲノム・遺伝子情報のWebツール
- ② 遺伝子変異・SNP情報のWebツール
- ③ 疾患感受性遺伝子情報・解析結果のWebツール
- ④ エピゲノム情報のWebツール
- ⑤ 創薬情報のWebツール

③-1: GWAS catalog



GWAS Catalog

Home

Diagram

Submit

Download

Documentation

About

EMBL-EBI



National Human Genome Research Institute



GWAS Catalog

The NHGRI-EBI Catalog of human genome-wide association studies

Search the catalog



Examples: breast carcinoma, rs7329174, Yao, 2q37.1, HBS1L, 6:16000000-25000000

Download

Download a full copy of the GWAS Catalog in spreadsheet format as well as current and older versions of the GWAS diagram in SVG format.

Summary statistics

Documentation and access to full summary statistics for GWAS Catalog studies where available.

Submit

Submit summary statistics to GWAS Catalog.

Documentation

Including FAQs, our curation process, training materials

Diagram

Explore an interactive visualisation of all SNP-trait

Ancestry

An introduction to our ancestry curation process

<https://www.ebi.ac.uk/gwas/>

- GWAS結果(疾患名・遺伝子名・SNP名・論文名)のアーカイブサイトです。
- "height"と入力してみましょう。

③-1: GWAS catalog

Refine search results

P Publications 34

T Traits 18

Catalog stats

- Last data release on 2022-07-30
- 5876 publications
- 220322 SNPs
- 402121 associations
- Genome assembly GRCh38.p13
- dbSNP Build 154
- Ensembl Build 107

Search results for *height*

T body height EFO_0004339

The distance from the sole to the crown of the head with body standing on a flat surface and fully extended.

Associations 7102 Studies 86

T sitting height measurement EFO_0011011

Quantification of some aspect of height, when assuming a sitting position.

Associations 11 Studies 5

T infant body height EFO_0006785

The distance from the sole to the crown of the head of an infant, which can be used as a proxy for skeletal growth in early life

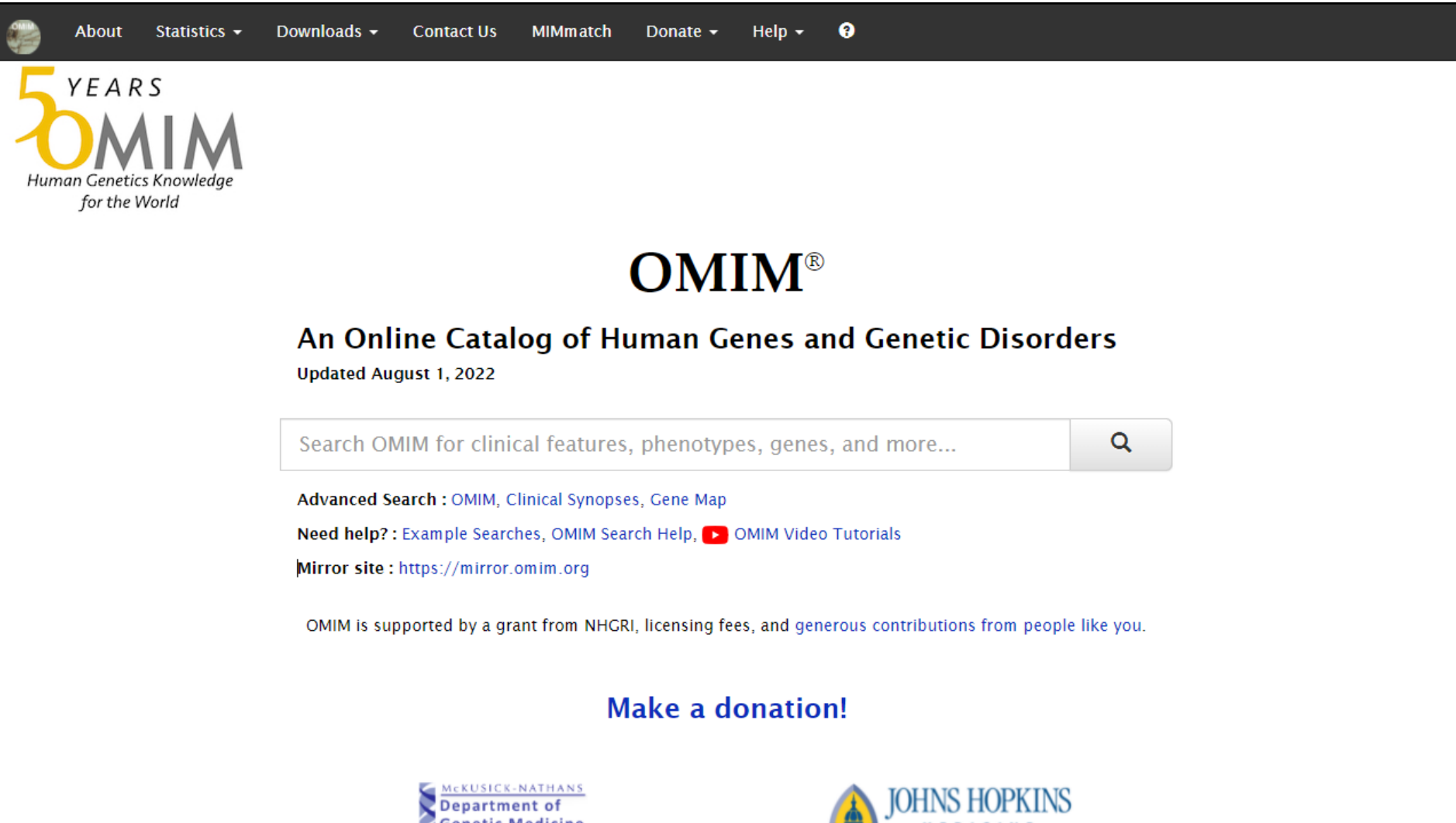
Associations 15 Studies 1

T sitting height ratio EFO_0007118

<https://www.ebi.ac.uk/gwas/>

- 身長のGWAS結果(遺伝子名・SNP名・論文名)の一覧が表示されます。
- 遺伝子名、SNP名、論文名でも検索可能です。

③-2: OMIM (Online Mendelian Inheritance in Man)



OMIM®

An Online Catalog of Human Genes and Genetic Disorders

Updated August 1, 2022

Search OMIM for clinical features, phenotypes, genes, and more...

Advanced Search : [OMIM](#), [Clinical Synopses](#), [Gene Map](#)

Need help? : [Example Searches](#), [OMIM Search Help](#), [OMIM Video Tutorials](#)

Mirror site : <https://mirror.omim.org>

OMIM is supported by a grant from NHGRI, licensing fees, and generous contributions from people like you.

[Make a donation!](#)

McKUSICK-NATHANS
Department of
Genetic Medicine

JOHNS HOPKINS

<http://www.omim.org/>

- 希少難病疾患を中心に、原因遺伝子情報を集約したデータベースです。
- “ALS”と入力してみましょう。

③-2:OMIM (Online Mendelian Inheritance in Man)

The screenshot shows the OMIM website interface. At the top, there is a navigation bar with links for 'About', 'Statistics', 'Downloads', 'Contact Us', 'MIMmatch', 'Donate', 'Help', and a question mark icon. Below the navigation bar is a search bar containing the text 'ALS'. To the right of the search bar are buttons for 'View Results as: Gene Map Table' and 'Clinical Synopsis', and a 'Display: Highlights' checkbox. The search results are listed below, showing 206 entries. The first six entries are:

- 1: # 105500. AMYOTROPHIC LATERAL SCLEROSIS-PARKINSONISM/DEMENTIA COMPLEX 1
Cytogenetic location: 15q21.2
Matching terms: als
▶ Phenotype-Gene Relationships ▶ ICD+ ▶ Links
- 2: # 205100. AMYOTROPHIC LATERAL SCLEROSIS 2, JUVENILE; ALS2
Cytogenetic location: 2q33.1
Matching terms: als
▶ Phenotype-Gene Relationships ▶ Phenotypic Series ▶ ICD+ ▶ Links
- 3: % 205200. AMYOTROPHIC LATERAL SCLEROSIS, JUVENILE, WITH DEMENTIA
Matching terms: als
▶ Phenotypic Series ▶ ICD+ ▶ Links
- 4: * 601489. INSULIN-LIKE GROWTH FACTOR-BINDING PROTEIN, ACID-LABILE SUBUNIT; IGFALS
Cytogenetic location: 16p13.3, Genomic coordinates (GRCh38): 16:1,790,413-1,794,908
Matching terms: als
▶ Gene-Phenotype Relationships ▶ Links
- 5: # 105400. AMYOTROPHIC LATERAL SCLEROSIS 1; ALS1
AMYOTROPHIC LATERAL SCLEROSIS 1, AUTOSOMAL RECESSIVE, INCLUDED
Cytogenetic locations: 2p13.1, 12q13.12, 21q22.11, 22q12.2
Matching terms: als
▶ Phenotype-Gene Relationships ▶ Phenotypic Series ▶ ICD+ ▶ Links
- 6: * 147450. SUPEROXIDE DISMUTASE 1; SOD1
Cytogenetic location: 21q22.11, Genomic coordinates (GRCh38): 21:31,659,693-31,668,931

<http://www.omim.org/>

・専門家によって書かれた、ALS(筋萎縮性側索硬化症)に関する詳細な説明と、感受性遺伝子や領域の情報が提供されます。

③-3: COSMIC (Catalogue of Somatic Mutations in Cancer)

COSMIC
Catalogue Of Somatic Mutations In Cancer

Projects ▾ Data ▾ Tools ▾ News ▾ Help ▾ About ▾ Genome Version ▾ Search COSMIC... **SEARCH** Login ▾

COSMIC v96, released 31-MAY-22

COSMIC, the Catalogue Of Somatic Mutations In Cancer, is the world's largest and most comprehensive resource for exploring the impact of somatic mutations in human cancer.

Start using COSMIC by searching for a gene, cancer type, mutation, etc. below.

eg Braf, COLO-829, Carcinoma, V600E, BRCA-UK, Campbell **SEARCH**

Projects

COSMIC is divided into several distinct projects, each presenting a separate dataset or view of our data:

- COSMIC**
The core of COSMIC, an expert-curated database of somatic mutations
- Cell Lines Project**
Mutation profiles of over 1,000 cell lines used in cancer research
- COSMIC-3D**
An interactive view of cancer mutations in the context of 3D structures
- Cancer Gene Census**
A catalogue of genes with mutations that are causally implicated in

COSMIC News

[Follow @cosmic_sanger](#)

- Copy Number Signatures: A scalable research and clinical tool?**
Copy Number Signatures are the latest addition to COSMIC Mutational Signatures, we caught up with one of the leads, Dr Ludmil Alexandrov, to discuss the key findings, utility of the data, and hopes for the future. [More...](#)
- What are the emerging trends in cancer research? Our five key-takeaways from AACR-2022**
Read about the five emerging trends we took away from our time at AACR-2022 [More...](#)
- Closing the care gap for rare cancers: Three examples in COSMIC**
Closing the care gap through COSMIC's curation of rare cancers. [More...](#)

Tools

- [Cancer Browser](#) — browse COSMIC data by tissue type and histology
- [Genome Browser](#) — browse the human genome with COSMIC annotations
- [GA4GH Beacon](#) — access COSMIC data through the [GA4GH Beacon Project](#)

<http://cancer.sanger.ac.uk/cosmic>

・がん体細胞変異情報が蓄積されたカタログデータベースです。

③-4:MR-base (Mendelian Randomization-base)



2-sample Mendelian Randomisation



Home MR-Base web app R package ↗ MRC IEU OpenGWAS PheWAS Publications

MR-base is a database and analytical platform for Mendelian randomization being developed by the [MRC Integrative Epidemiology Unit](#) at the University of Bristol.

You can either use the web application or our [TwoSampleMR R package](#).

Data are also available through the [MRC IEU OpenGWAS database](#).

[Launch MR-Base webapp](#)

[R package](#)

[OpenGWAS database](#)

Note - by clicking the "Launch MR-Base webapp" button you consent to the use of a cookie which enables us to ensure you have consented to the terms and conditions of data access. Information about how to control or delete cookies can be found at www.aboutcookies.org

MR-Base paper published

The MR-Base paper has now been published in eLife. See the [publications page](#) for details.

Telomeres paper published

Our paper reporting Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases has been published in *Jama Oncology*. See the [publications page](#) to access supporting data.

MR-Base Citation

<http://www.mrbase.org/>

• **M**endelian **R**andomization (**MR**): 複数のGWAS結果を比較し、遺伝的疾患リスクの**因果関係**を検討する手法。

③-4:MR-base (Mendelian Randomization-base)



2-sample Mendelian Randomisation



Home

MR-Base web app

R package ↗

MRC IEU OpenGWAS

PheWAS

Publications

MR-base is a database and analytical platform for Mendelian randomization being developed by the [MRC Integrative Epidemiology Unit](#) at the University of Bristol.

You can either use the web application or our [TwoSampleMR R package](#).

Data are also available through the [MRC IEU OpenGWAS database](#).

Launch MR-Base webapp

R package

OpenGWAS database

Note - by clicking the "Launch MR-Base webapp" button you consent to the use of a cookie which enables us to ensure you have consented to the terms and conditions of data access. Information about how to control or delete cookies can be found at www.aboutcookies.org

MR-Base paper published

The MR-Base paper has now been published in eLife. See the [publications page](#) for details.

Telomeres paper published

Our paper reporting Association Between Telomere Length and Risk of Cancer and Non-Neoplastic Diseases has been published in *Jama Oncology*. See the [publications page](#) to access supporting data.

MR-Base Citation

<http://www.mrbase.org/>

•MRの実行だけでなく、数多くのGWAS結果(ゲノムワイドSNPの統計量)をダウンロードすることができます。

③-5: UKbiobank (Pan-UK Biobank)



Pan-UK Biobank

About News Downloads Phenotypes Team Contact

Pan-UK Biobank

Pan-ancestry genetic analysis of the UK Biobank

The UK Biobank is a collection of a half million individuals with paired genetic and phenotype information that has been enormously valuable in studies of genetic etiology for common diseases and traits. However, most genome-wide analyses of this dataset use only the European ancestry individuals. Analyzing a more inclusive and diverse dataset increases power

<https://pan.ukbb.broadinstitute.org/>

- UKバイオバンクにおけるGWAS結果をダウンロードできるサイトです。
- Harvard大学のBenjamin Neale博士の研究室が運営しています。

③-6: Gene ATLAS



[Home](#) [Search](#) [Trait Info](#) [Downloads](#) [Correlations](#) [FAQ](#)

Gene ATLAS is a large database of associations between hundreds of traits and millions of variants using the UK Biobank cohort.

The associations have been computed using 452,264 UK Biobank White British individuals. To avoid having to remove the large amount of related individuals present on the study, the associations have been computed using Mixed Linear Models in a large supercomputer using **DISSECT**. The objective of the current database is to benefit the research community by making a searchable atlas of genetic associations that help researchers to query associations results in an easy way, without the need to incur in the high computational costs required to analyze the UK Biobank large cohort.

452264

Individuals

778

Traits

30

Million Variants

<http://geneatlas.roslin.ed.ac.uk/>

- UKバイオバンクにおけるGWAS結果をダウンロードできるサイトです。
- Edinburgh大学のAlbert Tenesa博士の研究室が運営しています。

③-7: Pheweb.jp

Search for a variant, gene, or phenot

andom Downloads About

検索Box(形質・遺伝子・SNP)

BioBank Japan PheWeb (PheWeb.jp)

Search for a variant, gene, or phenotype

Example queries: **Type 2 diabetes (T2D)** **PCSK9** **rs671** ← **検索例**

This website releases genome-wide association study (GWAS) summary statistics of **the BioBank Japan Project (BBJ)**. We provide GWAS results in the Japanese population (mainly from BBJ) using the PheWeb platform, with public access to the full summary statistics.

BBJ is a prospective genome biobank that collaboratively collected DNA and serum samples from 12 medical institutions in Japan, managed by **the Institute of Medical Science, the University of Tokyo**. BBJ has recruited approximately 260,000 participants, mainly of Japanese ancestry. All study participants had been diagnosed with one or more of 47 target diseases. RIKEN Center for Integrative Medical Sciences contributed to genotyping of the BBJ samples.

News

March 12, 2022: cis-miRNA-eQTL results from **Sonehara, K. & Sakaue, S., et al. (2022)**.
Raw data are available for download on **the NBDC website**

<https://pheweb.jp/>

• **バイオバンク・ジャパン(BBJ)を中心に、日本人集団における多彩な形質のGWASの全SNPの結果がダウンロードできるサイトです。**

③-7:Pheweb.jp



Search for a variant, gene, or phenotype

Phenotypes Top Hits Random Downloads About

T2D: Type 2 diabetes

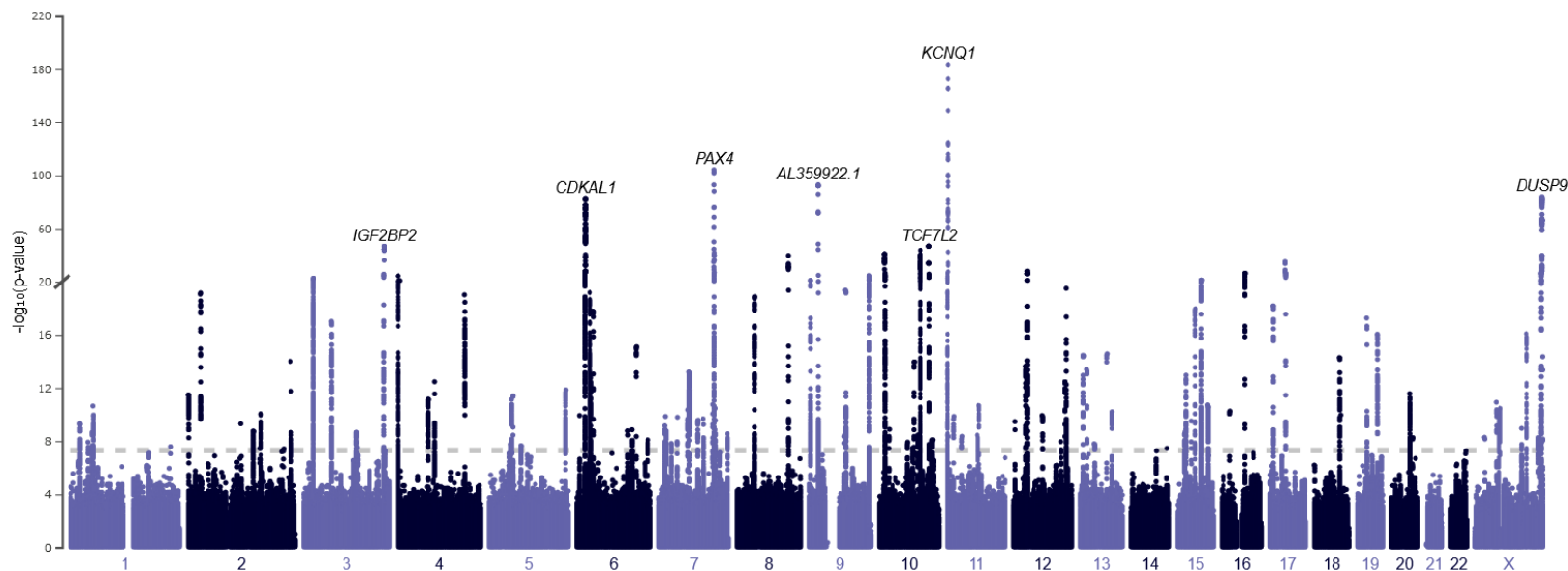
45383 cases, 132032 controls

Category: ICD10 L

Citation: Sakaue, S. & Kanai, M., et al. [A global atlas of genetic associations of 220 deep phenotypes](#). medRxiv (2020)

Download summary statistics

Manhattan QQ



Top Loci:

Search: "TCF7L2", "rs1861867", etc

167 total variants

<https://pheweb.jp/>

- 形質名、遺伝子名、SNP名で検索すると、結果が図示されます。
- 形質名“Type 2 diabetes”を入力してみましょう。

③-7: Pheweb.jp



Search for a variant, gene, or phenotype

Phenotypes Top Hits Random Downloads About

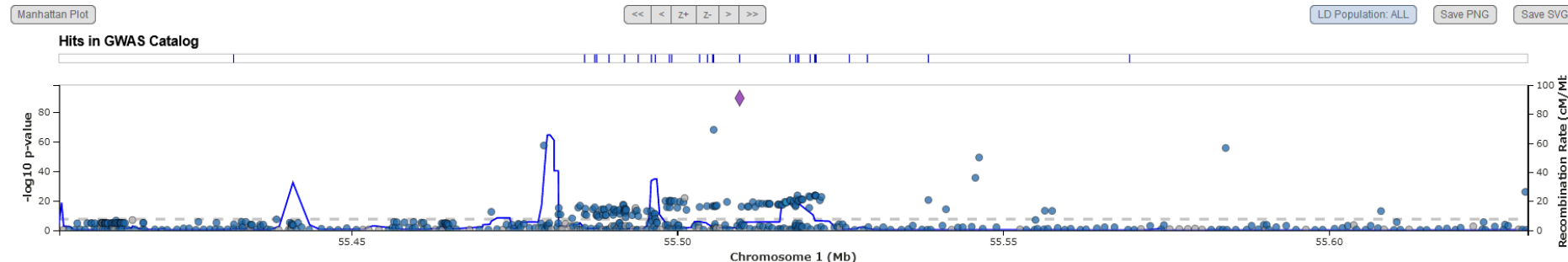
PCSK9

Phenotypes with the most-significant associations for this locus:

| Top p-value in gene | Phenotype |
|---------------------|---------------------------------------|
| 8.2e-90 | Total cholesterol |
| 3.6e-70 | LDL-cholesterol |
| 2.9e-51 | HMG CoA reductase inhibitors |
| 7.1e-11 | Salicylic acid and derivatives |
| 9.3e-10 | Myocardial infarction |
| 4.2e-9 | Vasodilators used in cardiac diseases |
| 1.6e-8 | Antithrombotic agents |
| 1.9e-8 | Unstable angina pectoris |

135808 samples

Category: Metabolic



<https://pheweb.jp/>

- 形質名、遺伝子名、SNP名で検索すると、結果が図示されます。
- 遺伝子名“PCSK9”を入力してみましょう。

③-7: Pheweb.jp



Search for a variant, gene, or phenotype

Phenotypes Top Hits Random Downloads About

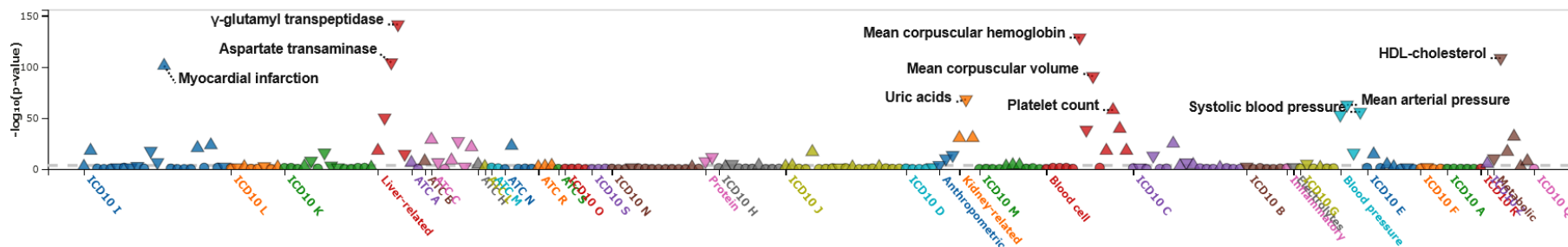
12 : 112,241,766 G / A (rs671)

Nearest gene: *ALDH2*

AF ranges from 0.24 to 0.25

View on UCSC , GWAS Catalog , GTEx , LitVar , dbSNP , gnomAD , PubMed (219 results) , Clinvar

Save PNG Save SVG



Search... "427.21", "Diabetes", etc.

218 total codes

| Category | Phenotype | P-value | Effect Size (se) | Number of samples |
|----------------|-----------------------------|----------|------------------|-------------------|
| Liver-related | γ-glutamyl transpeptidase | ≤1e-320 | -0.17 (0.0040) | 133471 |
| Blood cell | Mean corpuscular hemoglobin | 3.6e-129 | -0.10 (0.0042) | 128028 |
| Metabolic | HDL-cholesterol | 3.9e-109 | -0.12 (0.0055) | 74970 |
| Liver-related | Aspartate transaminase | 7.9e-105 | -0.088 (0.0041) | 150068 |
| ICD10 I | Myocardial infarction | 9.5e-102 | 0.33 (0.016) | 14992 / 146214 |
| Blood cell | Mean corpuscular volume | 2.0e-91 | -0.085 (0.0042) | 129832 |
| Kidney-related | Uric acids | 7.5e-69 | -0.072 (0.0041) | 129405 |

<https://pheweb.jp/>

• 形質名、遺伝子名、SNP名で検索すると、結果が図示されます。

• SNP名“rs671”を入力してみましょう。



Search for a variant, gene, or phenotype

Phenotypes

Top Hits

Random

Downloads

About

Downloads

The GWAS results on this website are from the following studies:

A global atlas of genetic associations of 220 deep phenotypes
Sakaue, S. & Kanai, M., et al, *medRxiv* (2020)

Genetic determinants of risk in autoimmune pulmonary alveolar proteinosis
Sakaue, S., et al, *Nat. Commun.* **12** 1032 (2021)

Genetics of rheumatoid arthritis contributes to biology and drug discovery
Okada, Y., et al, *Nature* **506** 376–381 (2014)

When using these results, please cite the relevant studies which appear on each phenotype page.

Sakaue, S. & Kanai, M., et al. (2020) ▾

Sakaue, S., et al. (2021)

Okada, Y., et al. (2014)

GWAS summary statistics of BioBank Japan (BBJ)

Citation: Sakaue, S. & Kanai, M., et al. A global atlas of genetic associations of 220 deep phenotypes. *medRxiv* (2020).

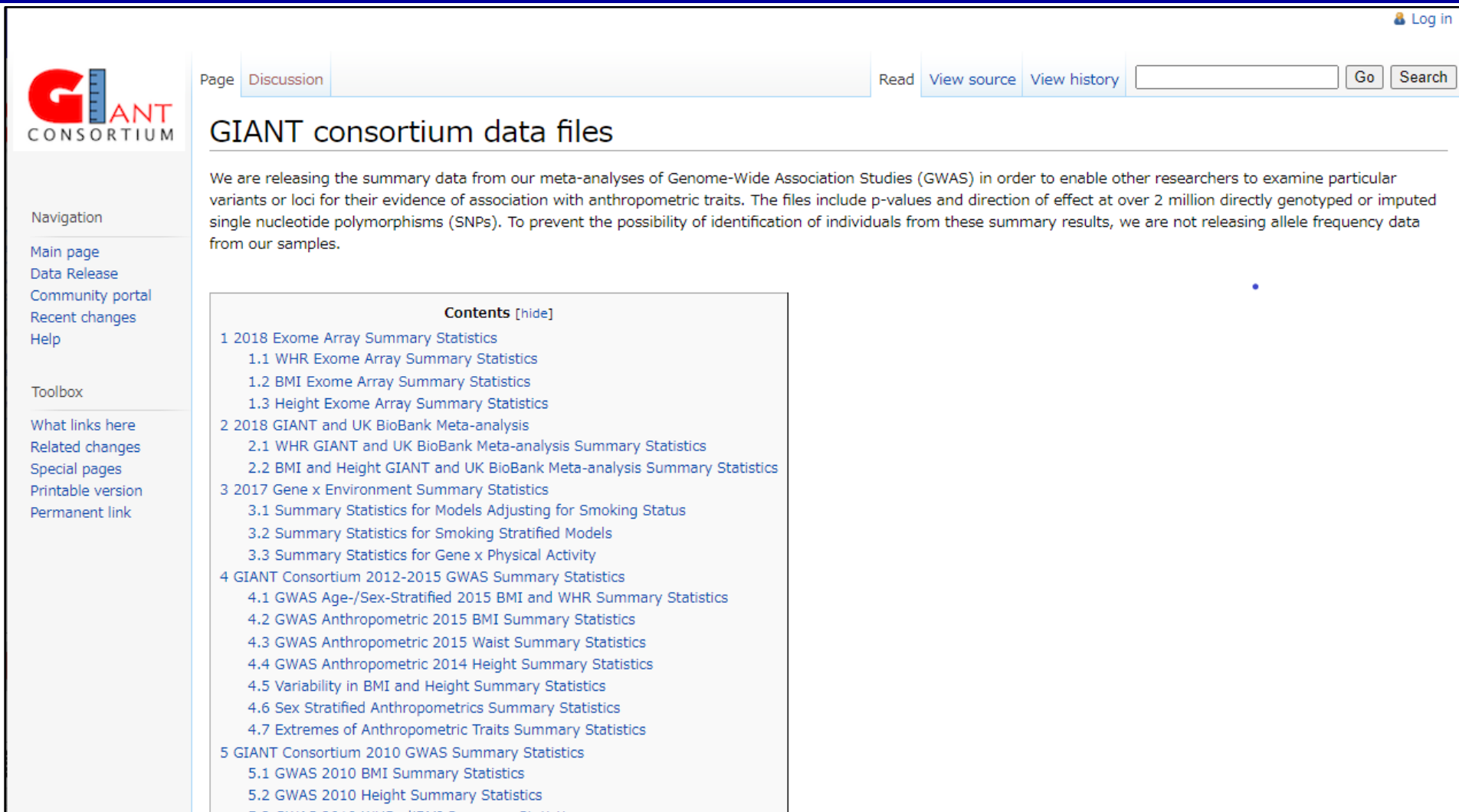
[Link to wget commands for all the files](#)

| Category | Phenotype | Cohort | No. samples | No. cases | No. controls | Download link |
|----------|----------------------|--------|-------------|-----------|--------------|--------------------------|
| ICD10 A | Diphtheria infection | BBJ | 170,788 | 541 | 170,247 | Download |
| ICD10 A | Dysentery | BBJ | 178,482 | 411 | 178,071 | Download |

<https://pheweb.jp/>

• “Downloads” ページからは、全GWASの全SNPの結果をダウンロードすることができます。

③-8:GIANT consortium



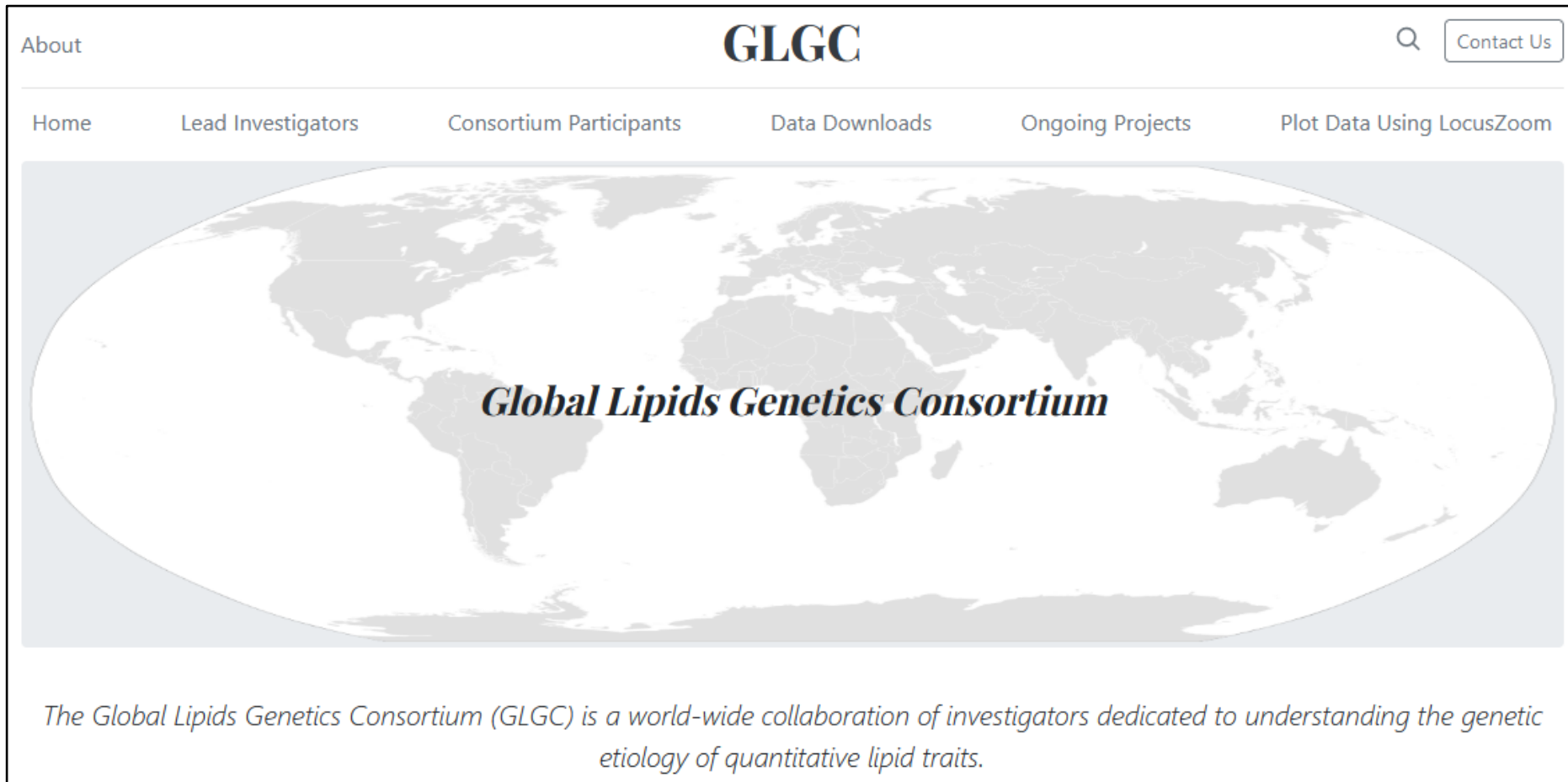
The screenshot shows the website for the GIANT Consortium data files. The page title is "GIANT consortium data files". The main content area contains a paragraph explaining the release of summary data from meta-analyses of Genome-Wide Association Studies (GWAS) for anthropometric traits. Below the text is a table of contents with the following items:

- 1 2018 Exome Array Summary Statistics
 - 1.1 WHR Exome Array Summary Statistics
 - 1.2 BMI Exome Array Summary Statistics
 - 1.3 Height Exome Array Summary Statistics
- 2 2018 GIANT and UK BioBank Meta-analysis
 - 2.1 WHR GIANT and UK BioBank Meta-analysis Summary Statistics
 - 2.2 BMI and Height GIANT and UK BioBank Meta-analysis Summary Statistics
- 3 2017 Gene x Environment Summary Statistics
 - 3.1 Summary Statistics for Models Adjusting for Smoking Status
 - 3.2 Summary Statistics for Smoking Stratified Models
 - 3.3 Summary Statistics for Gene x Physical Activity
- 4 GIANT Consortium 2012-2015 GWAS Summary Statistics
 - 4.1 GWAS Age-/Sex-Stratified 2015 BMI and WHR Summary Statistics
 - 4.2 GWAS Anthropometric 2015 BMI Summary Statistics
 - 4.3 GWAS Anthropometric 2015 Waist Summary Statistics
 - 4.4 GWAS Anthropometric 2014 Height Summary Statistics
 - 4.5 Variability in BMI and Height Summary Statistics
 - 4.6 Sex Stratified Anthropometrics Summary Statistics
 - 4.7 Extremes of Anthropometric Traits Summary Statistics
- 5 GIANT Consortium 2010 GWAS Summary Statistics
 - 5.1 GWAS 2010 BMI Summary Statistics
 - 5.2 GWAS 2010 Height Summary Statistics
 - 5.3 GWAS 2010 WHR/BMI Summary Statistics

http://portals.broadinstitute.org/collaboration/giant/index.php/GIANT_consortium_data_files

•身長/肥満GWASの全SNPの結果がダウンロードできるサイトです。 47

③-9: Global Lipids Genetics Consortium

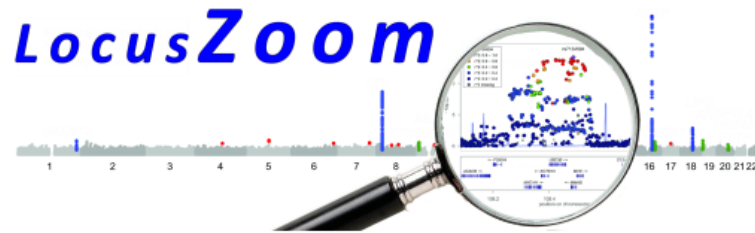


The screenshot shows the homepage of the Global Lipids Genetics Consortium (GLGC). At the top, there is a navigation bar with the following links: "About", "Home", "Lead Investigators", "Consortium Participants", "Data Downloads", "Ongoing Projects", and "Plot Data Using LocusZoom". The "GLGC" logo is prominently displayed in the center of the navigation bar. To the right of the logo, there is a search icon and a "Contact Us" button. Below the navigation bar, there is a large banner image featuring a world map. Overlaid on the map is the text "Global Lipids Genetics Consortium" in a bold, italicized font. Below the banner, there is a paragraph of text: "The Global Lipids Genetics Consortium (GLGC) is a world-wide collaboration of investigators dedicated to understanding the genetic etiology of quantitative lipid traits."

<http://lipidgenetics.org/>

•脂質GWASの全SNPの結果がダウンロードできるサイトです。

③-10:Locus Zoom



LocusZoom is a suite of tools to provide fast visualization of GWAS results for research and publication.

Original LocusZoom (R/Python) is ideal for batch generation of static plots.

LocusZoom.js (JavaScript) aims to make LocusZoom plots interactive and scriptable.

Interactive Plots with LocusZoom.js



MY.LOCUSZOOM.ORG

UPLOAD, ANALYZE, AND SHARE



LOCALZOOM

EXPLORE WITHOUT UPLOADING

Legacy Services (not actively maintained)

SINGLE PLOT

YOUR DATA - ORIGINAL LOCUSZOOM

BATCH PLOT WITH HITSPEC

YOUR DATA - ORIGINAL LOCUSZOOM

INTERACTIVE PLOT

PUBLISHED GWAS - LOCUSZOOM.JS

News

Planned Portaldev API downtime ~ November 13, 2020

Links

USER SURVEY

<http://locuszoom.sph.umich.edu/locuszoom/>

•GWAS解析結果の、領域内SNP P値の図を描くことができるサイトです。

Webツール入門

- ① ゲノム・遺伝子情報のWebツール
- ② 遺伝子変異・SNP情報のWebツール
- ③ 疾患感受性遺伝子情報・解析結果のWebツール
- ④ エピゲノム情報のWebツール
- ⑤ 創薬情報のWebツール

④-1: GEO database

NCBI Resources How To Sign in to NCBI

GEO Home Documentation Query & Browse Email GEO

Gene Expression Omnibus

GEO is a public functional genomics data repository supporting MIAME-compliant data submissions. Array- and sequence-based data are accepted. Tools are provided to help users query and download experiments and curated gene expression profiles.

Keyword or GEO Accession Search

Getting Started

- Overview
- FAQ
- About GEO DataSets
- About GEO Profiles
- About GEO2R Analysis
- How to Construct a Query
- How to Download Data

Tools

- Search for Studies at GEO DataSets
- Search for Gene Expression at GEO Profiles
- Search GEO Documentation
- Analyze a Study with GEO2R
- Studies with Genome Data Viewer Tracks
- Programmatic Access
- FTP Site
- ENCODE Data Listings and Tracks

Browse Content

| | |
|--------------------|---------|
| Repository Browser | |
| DataSets: | 4348 |
| Series: | 180737 |
| Platforms: | 24185 |
| Samples: | 5188604 |

Information for Submitters

| | | |
|-----------------|-----------------------|---------------------------|
| Login to Submit | Submission Guidelines | MIAME Standards |
| | Update Guidelines | Citing and Linking to GEO |
| | | Guidelines for Reviewers |

<http://www.ncbi.nlm.nih.gov/geo/>

• 遺伝子発現データベースサイトです。論文投稿時には発現データ登録が義務づけられている例が多いです。“GSE45878”と入力してみま⁵¹しょう。

④-1: GEO database

The screenshot shows the NCBI GEO database interface. At the top, there are logos for NCBI and GEO (Gene Expression Omnibus). Below the logos is a navigation bar with links for HOME, SEARCH, SITE MAP, GEO Publications, FAQ, MIAME, and Email GEO. The main content area displays the accession number GSE45878. A search bar at the top of the content area shows 'Scope: Self', 'Format: HTML', 'Amount: Quick', and 'GEO accession: GSE45878'. Below this, the series title 'Series GSE45878' is shown, along with a link to 'Query DataSets for GSE45878'. The main text area contains the following information:

Status Public on Apr 09, 2013
Title Affymetrix expression data from GTEx
Organism [Homo sapiens](#)
Experiment type Expression profiling by array
Summary The Genotype-Tissue Expression (GTEx) project is a collaborative effort that aims to identify correlations between genotype and tissue-specific gene expression levels that will help identify regions of the genome that influence whether and how much a gene is expressed. GTEx is funded through the Common Fund, and managed by the NIH Office of the Director in partnership with the National Human Genome Research Institute, National Institute of Mental Health, the National Cancer Institute, the National Center for Biotechnology Information at the National Library of Medicine, the National Heart, Lung and Blood Institute, the National Institute on Drug Abuse, and the National Institute of Neurological Diseases and Stroke, all part of NIH. This series of 837 samples represents multiple tissues collected from 102 GTEx donors and 1 control cell line. In total, 30 tissue sites are represented including Adipose, Artery, Heart, Lung, Whole Blood, Muscle, Skin, and 11 brain subregions. RNA-seq expression data, robust clinical data, pathological annotations, and genotypes are also available for these samples from dbGaP (http://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/study.cgi?study_id=phs000424.v2.p1) and the GTEx portal (www.broadinstitute.org/gtex). While GTEx is no longer generating Affymetrix expression data, donor enrollment continues and is expected to reach 1,000 by the end of 2015. Updates to the GTEx data in dbGaP and the GTEx Portal will be made periodically.
contributor: GTEx Laboratory, Data Analysis, and Coordinating Center (LDACC)
contributor: The Broad Institute of MIT and Harvard (LDACC PIs: Kristin Ardlie and Gaddy Getz)

Overall design GTEx samples are collected from deceased donors at low post-mortem intervals and preserved in PAXgene fixative prior to DNA and RNA extraction.

<http://www.ncbi.nlm.nih.gov/geo/>

- ④-2で解析されているGTExプロジェクトで得られた遺伝子発現データが公開されています。ダウンロード可能です。

④-2:GTEx Portal

GTEx Portal

About GTEx Publications Access Biospecimens FAQs Contact

Home Downloads Expression Single Cell QTL IGV Browser Tissues & Histology Documentation Search Gene or SNP ID... Sign In

The GTEx and HuBMAP teams would like your feedback on the integration of the Exploration User Interface. Please answer our survey here: <https://forms.gle/FYHSTHLjxCir6piCa>

2022-07-21
GTEX Samples Mapped to HuBMAP Common Coordinate Framework
We have spatially mapped GTEx samples from 16 tissue sites to the [HuBMAP Common Coordinate Framework](#). We have embedded the HuBMAP

Resource Overview

Current Release (V8)
Tissue & Sample Statistics
Tissue Sampling Info (Anatomogram)
Access & Download Data
Release History
How to cite GTEx?

The Genotype-Tissue Expression (GTEx) project is an ongoing effort to build a comprehensive public resource to study tissue-specific gene expression and regulation. Samples were collected from 54 non-diseased tissue sites across nearly 1000 individuals, primarily for molecular assays including WGS, WES, and RNA-Seq. Remaining samples are available from the GTEx Biobank. The GTEx Portal provides open access to data including gene expression,

Explore GTEx

Browse
By gene ID
By variant or rs ID
By Tissue
Histology Viewer

Single Cell
Data Overview
Multi-Gene Single Cell Query

Browse and search all data by gene
Browse and search all data by variant
Browse and search all data by tissue
Browse and search GTEx histology images
Learn more about available single cell data
Browse and search single cell expression by gene and tissue

<http://www.gtexportal.org/home/>

• 900名の献体から得られた体内組織の遺伝子発現データが公開されたサイトです。“PADI2”と入力してみましょう。

④-2:GTEx Portal

The screenshot shows the GTEx Portal interface. At the top, there is a navigation bar with links for Home, Datasets, Expression, QTLs & Browsers, Sample Data, and Documentation. A search bar contains the text 'PADI2'. Below the navigation bar, there is a message asking users to take a survey. The main content area is titled 'Gene Page' and features a table with columns for Gene Symbol, Gencode ID, Entrez Gene ID, Location, Gene Description, and Actions. The table contains one entry for PADI2. Below the table, there are links to bulk tissue gene expression, single tissue expression, exon expression, and significant single-tissue eQTLs for PADI2.

Gene Page

Top

Bulk Tissue Expression

Single Cell Expression

Exon Expression

Single-Tissue eQTLs

Single-Tissue sQTLs

Single-Tissue ieQTLs

Single-Tissue isQTLs

Copy CSV

Search: Show 10 entries

| Gene Symbol | Gencode ID | Entrez Gene ID | Location | Gene Description | Actions |
|-------------|--------------------|----------------|--------------------------|---|----------------------------|
| PADI2 | ENSG00000117115.12 | 11240 | chr1:17066761-17119435:- | peptidyl arginine deiminase 2 [Source:HGNC Symbol;Acc:HGNC:18341] | IGV Browser, Ensembl, UCSC |

Showing 1 to 1 of 1 entries

First Previous 1 Next Last

Bulk tissue gene expression for PADI2 (ENSG00000117115.12)

Data Source: GTEx Analysis Release V8 (dbGaP Accession phs000424.v8.p2)

Data processing and normalization

Single tissue expression for PADI2 (ENSG00000117115.12)

Data Source: Single cell snRNA-seq pilot

Exon expression for PADI2 (ENSG00000117115.12)

Data Source: GTEx Analysis Release V8 (dbGaP Accession phs000424.v8.p2)

Significant Single-Tissue eQTLs for PADI2 (ENSG00000117115.12) in all tissues

Data Source: GTEx Analysis Release V8 (dbGaP Accession phs000424.v8.p2)

<http://www.gtexportal.org/home/>

•各組織におけるPADI2遺伝子の発現量と、SNPデータとの発現関連解析(eQTL解析)結果が得られます。

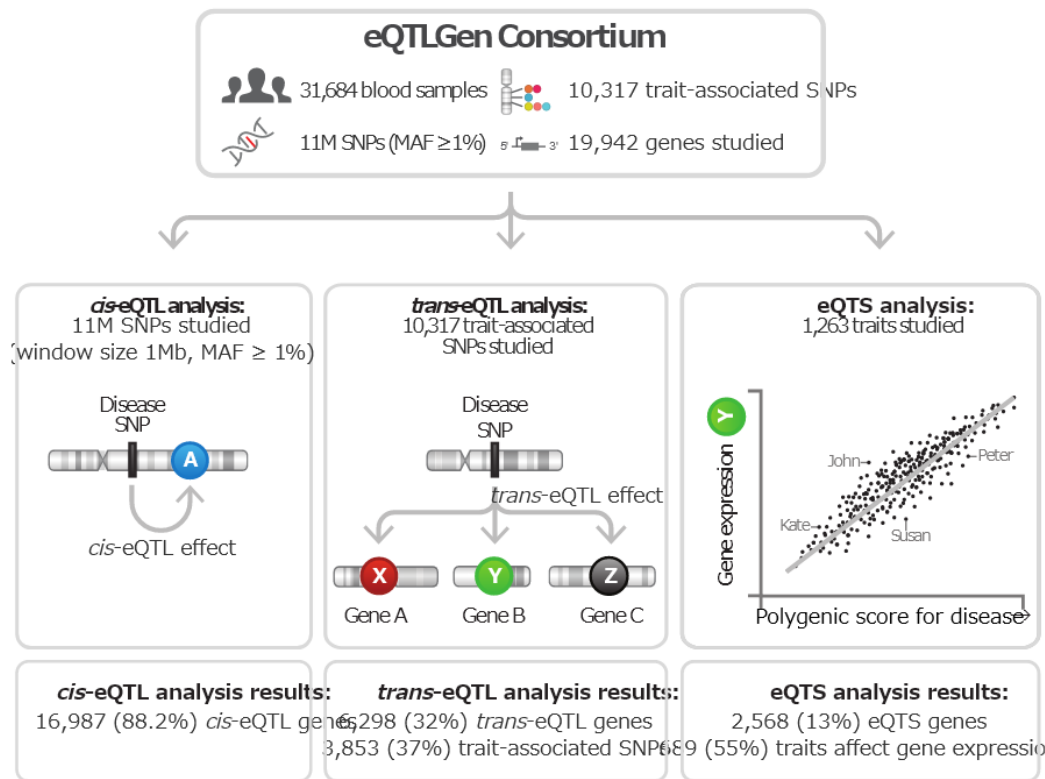
④-3:eQTLgen

eQTLGen

Cis-eQTLs Trans-eQTLs eQTS Replications Publications sc-eQTLGen

Welcome to the eQTLGen Consortium

The eQTLGen Consortium has been set up to identify the downstream consequences of trait-related genetic variants. The consortium incorporates 37 datasets, with a total of 31,684 individuals. You can find the [cis-eQTL](#), [trans-eQTL](#), [eQTS](#) and replication results from our forthcoming paper on this website.



<https://www.eqtlgen.org/index.html>

• eQTLgenコンソーシアムによる大規模末梢血遺伝子発現eQTL解析結果の公開サイトです。“Cis-eQTL”タブで“PADI2”と入力してみましょ⁵⁵う。

④-3:eQTLgen

cis-eQTLs

This page contains the *cis*-eQTL results. The statistically significant *cis*-eQTLs and SMR-prioritised genes for several traits are browsable, the other files can be downloaded.

Downloads

- Significant *cis*-eQTLs
- Full *cis*-eQTL summary statistics
- README
- Initial release Significant *cis*-eQTLs [deprecated]
- Initial release Full *cis*-eQTL summary statistics [deprecated]

Changelog

2019-12-23: *Cis*-eQTLs are now updated to have a 2-cohort filter: every *cis*-eQTL must be tested in at least 2 cohorts to be reported. The files now also include Bonferroni-adjusted p-values.

2018-10-19: Initial data release

Significant *cis*-eQTLs

Search:

Show entries

| P-value | SNP | | Chr | Pos [hg19] | Gene | | Symbol | Chr | Pos [hg19] | Z-score | Allele | | Nr Cohort | | |
|-------------|-----|-----------|-----|------------|----------|----|-----------------|-----|------------|----------|--------|----------|-----------|-------|----|
| | ↑↓ | ID | | | ↑↓ | ID | | | | | ↑↓ | Assessed | | Other | |
| 3.2717e-310 | ↑↓ | rs2076616 | ↑↓ | 1 | 17412501 | ↑↓ | ENSG00000117115 | ↑↓ | 1 | 17419602 | ↑↓ | -56.5846 | G | A | 37 |
| 3.2717e-310 | | rs2235927 | | 1 | 17394775 | | ENSG00000117115 | | 1 | 17419602 | | -46.2499 | A | G | 37 |

<https://www.eqtngen.org/index.html>

•PADI2遺伝子の発現量の個人差に影響を与える、周辺の遺伝子領域のSNPのリストが得られます。

④-4: ImmuNexUT

Immune Cell Gene Expression Atlas from the University of Tokyo

Browse by gene

Gene Symbol:

Browse by genetic variant

SNP ID:

FAQs
Publication
Cell. 2021; 184(11): 3006-3021

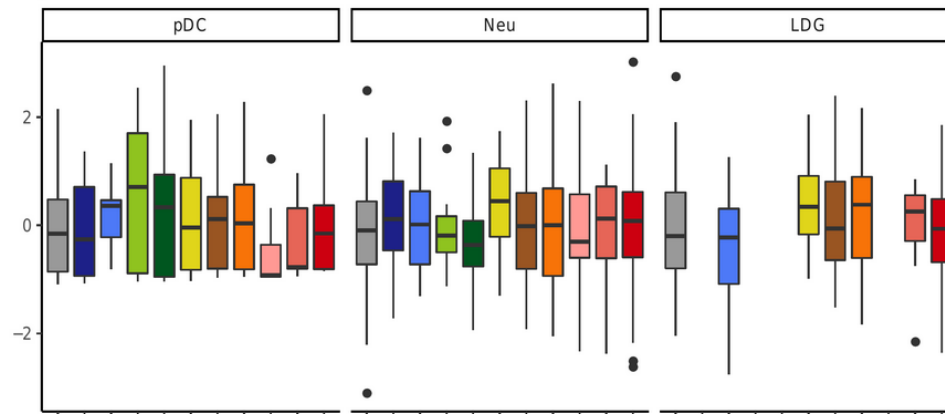
Department of Allergy and Rheumatology Graduate School of Medicine, The University of Tokyo
東京大学医学部附属病院 アレルギー・リウマチ内科
Free for academic or non-profit use; other users need a [Commercial license](#).

東京大学
THE UNIVERSITY OF TOKYO

<https://www.immunexut.org/>

- 日本人集団の免疫細胞特異的eQTLデータベースです。
- “PADI2”と入力してみましょう。

④-4: Human Genetic Variation Database



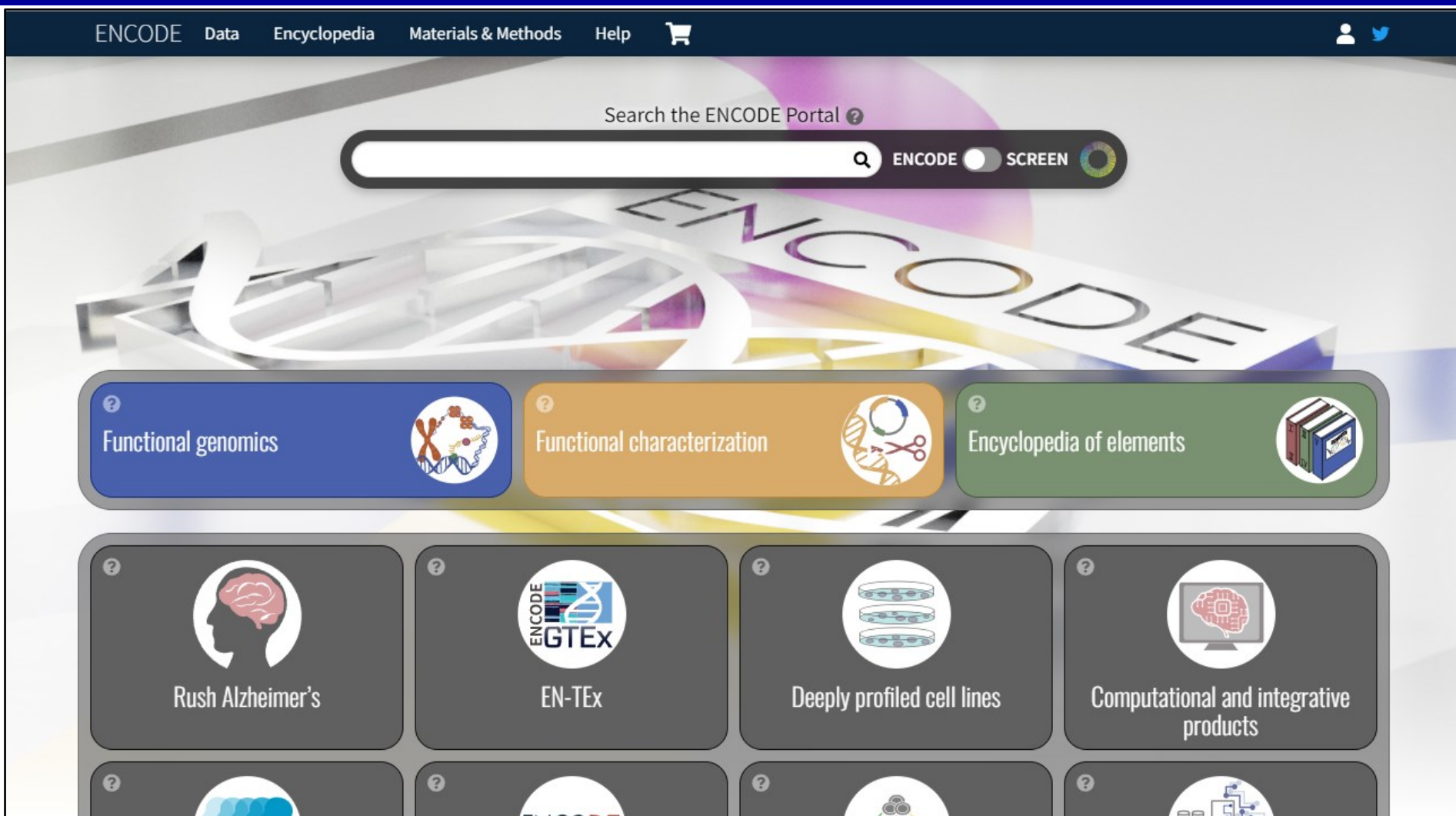
Significant eQTLs

| Gene Symbol | SNP ID | Cell Type | Chromosome | Position | eQTL p-value | eQTL effect beta | eQTL plot |
|-------------|----------------------------|-----------|------------|----------|--------------|------------------|-----------|
| PADI2 | rs2235912 | Neu | chr1 | 17098978 | 8.89475e-132 | 0.888588 | |
| PADI2 | rs2235913 | Neu | chr1 | 17098582 | 8.89475e-132 | 0.888588 | |
| PADI2 | rs2235914 | Neu | chr1 | 17098350 | 8.89475e-132 | 0.888588 | |
| PADI2 | rs10649306 | Neu | chr1 | 17097765 | 2.22036e-130 | 0.888127 | |
| PADI2 | rs11576552 | Neu | chr1 | 17096815 | 2.22036e-130 | 0.888127 | |

<https://www.immunexut.org/>

• PADI2遺伝子領域の遺伝子変異や、免疫細胞特異的なeQTL解析結果を見ることができます。

④-5: ENCODE (Encyclopedia of DNA Elements)



<https://www.encodeproject.org/>

・組織特異的エピゲノム情報を網羅するENCODEプロジェクトのサイトです。**"Data"→"Search by Region"**から**"PADI2"**と入力してみましょ⁵⁹う。

④-5: ENCODE (Encyclopedia of DNA Elements)

Region Search

Enter any one of human Gene name, Symbol, Synonyms, Gene ID, HGNC ID, coordinates, rsid, Ensemble ID

Homo sapiens ▾

Success

Search

Searched coordinates: **PADI2: (chr1:17066761-17119453) +/- 2kb**

Datasets

Genome Browser

Showing 25 of 2127

Assay ▾

Biosample term

| | |
|-------------------------|-----|
| K562 | 181 |
| GM12878 | 87 |
| head of caudate nucleus | 84 |
| HEK293 | 80 |
| middle frontal area 46 | 73 |

Target

| | |
|------|-----|
| CTCF | 246 |
|------|-----|

ATAC-seq in GM18508

Homo sapiens GM18508

Lab: Stephen Montgomery, Stanford

Project: ENCODE

Experiment

ENCSR357WQH

● released

📄 2 ● 1

ATAC-seq in DND-41

Homo sapiens DND-41

Experiment

ENCSR660WSB

<https://www.encodeproject.org/>

• PADI2 遺伝子領域における、ChIP-seq、DNase-seq などのエピゲノム情報が提供されます。

④-6: AMP (Accelerating Medicines Partnership)&ImmPort

U.S. Department of Health & Human Services



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ACCELERATING MEDICINES PARTNERSHIP® (AMP®)

Accelerating Medicines Partnership (AMP)

[Alzheimer's Disease](#)

[Common Metabolic Diseases](#)

[Parkinson's Disease](#)

[Rheumatoid Arthritis and Lupus](#)

[Schizophrenia](#)

[Type 2 Diabetes](#)

On this page

[AMP Partners](#)

[Timeline](#)

[Challenge](#)

[Impact](#)

[Governance](#)

Overview

Launched in 2014, the Accelerating Medicines Partnership® (AMP®) is a public-private partnership between the National Institutes of Health (NIH), the U.S. Food and Drug Administration (FDA), multiple biopharmaceutical and life science companies, non-profit and other organizations to transform the current



Related Information

[News: NIH, industry and non-profits join forces to speed validation of disease targets, February 4, 2014](#)

[Director's Blog: Introducing AMP: the Accelerating Medicines Partnership](#)

[AMP press conference](#)

[Multimedia](#)

[February 2014 Statement by the President](#)

[Foundation for the National Institutes of Health AMP Website](#)

<https://www.nih.gov/research-training/accelerating-medicines-partnership-amp>

• Accelerating Medicines Partnership (AMP) のサイトです。

④-6: AMP (Accelerating Medicines Partnership)&ImmPort



- Browse Shared Data using Faceted Search
- Download Data (Login Required)
- API Documentation
- Ask ImmPort Questions

News

22-June-22 ImmPort Data Release 44 is out! 38 new studies. For details please see the Data Release notes.

Enter text (or) Click Search to Explore ImmPort Shared Data e.g. influenza, COVID-19, SDY1 ...

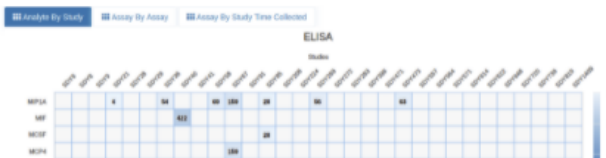
Search

Data Summary: Release 44, June 2022

Click on the counts with icon to visualize the count breakdown

| | | |
|-------------|---------------|-----------|
| Studies | Subjects | Diseases |
| 586 | 83513 | 130 |
| Experiments | Total Results | Lab Tests |
| 2146 | 6304584 | 1268118 |

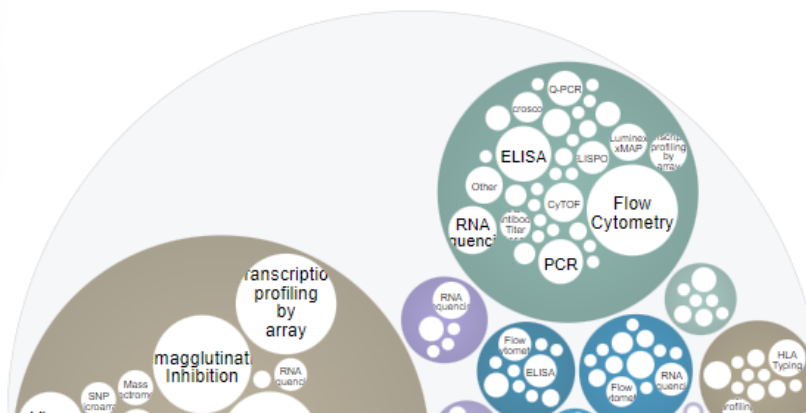
Matrix Summary - Analyte By Study



Bubble Summary: Research Focus by Assay Type

Click on a bubble to zoom in

Research Focus by Assay Type

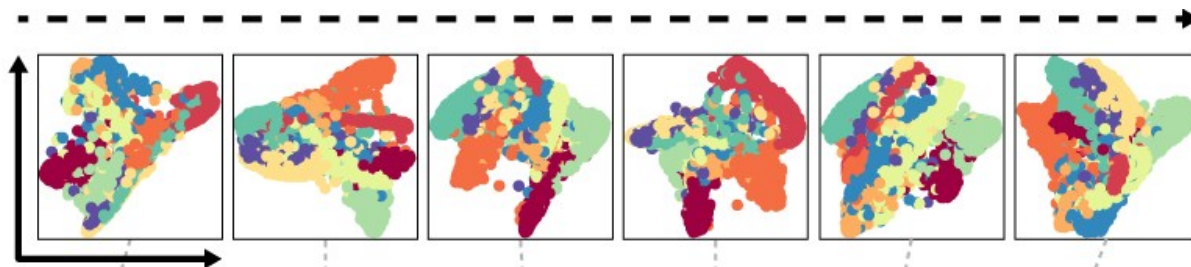


<https://www.immport.org/shared/home>

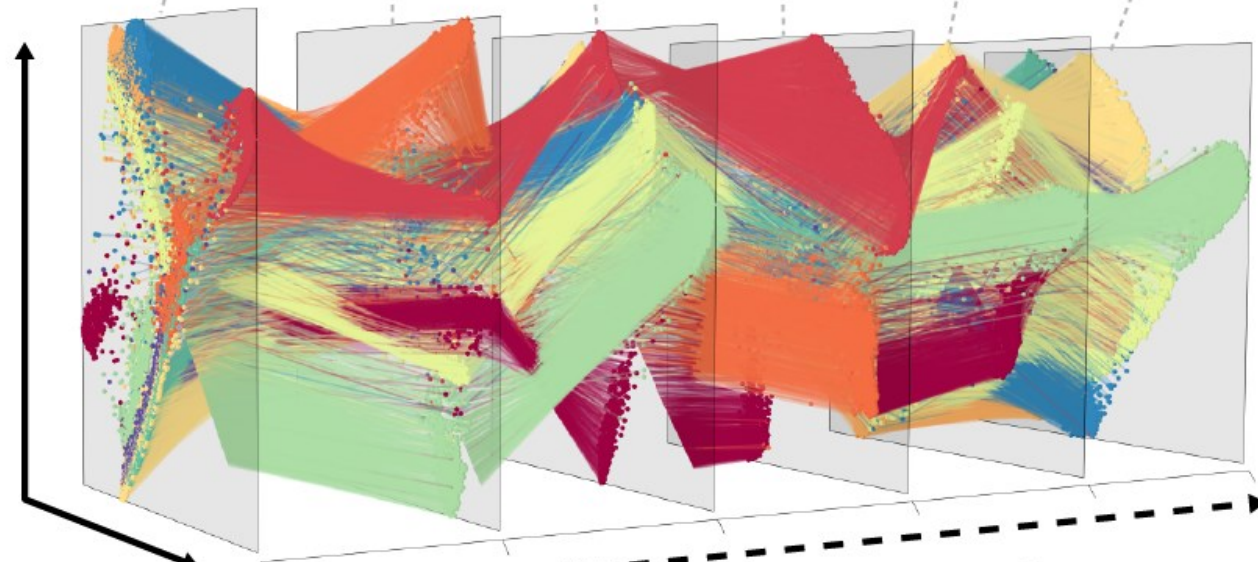
• Accelerating Medicines Partnership (AMP) によって構築されたエピゲノム・ゲノムデータをダウンロードできるサイトです。

④-7: Grimon (Graphical interface to visualize multi-omics networks)

2D visualization
in each layer



Grimon 3D
visualization



different data layers



<https://github.com/mkanai/grimon>

• 多次元のオミクス情報を次元圧縮し、感覚的に操作可能な3次元画像として可視化するGrimonのサイトです。

④-7: Grimon (Graphical interface to visualize multi-omics networks)

Product ▾ Team Enterprise Explore ▾ Marketplace Pricing ▾

Search / Sign in Sign up

mkanai / grimon Public

Notifications Fork 4 Star 16

<> Code Issues Pull requests Actions Projects Wiki Security Insights

master 1 branch 0 tags Go to file Code

| File/Folder | Commit Message | Time Ago |
|---------------|--|--------------|
| R | added point_alpha | 3 years ago |
| data | updated document | 4 years ago |
| man | add "optimized_point_mat" and "return_coordinates" | 4 years ago |
| src | updated v1.0.0 | 4 years ago |
| .Rbuildignore | first commit: v0.1.0 | 5 years ago |
| .gitignore | first commit: v0.1.0 | 5 years ago |
| DESCRIPTION | updated v1.0.0 | 4 years ago |
| LICENSE | Initial commit | 5 years ago |
| NAMESPACE | first commit: v0.1.0 | 5 years ago |
| README.md | format | 5 months ago |
| grimon.Rproj | first commit: v0.1.0 | 5 years ago |

About
R package for plotting 3D graphical visualization of multi-omics data.

Readme
GPL-3.0 license
16 stars
2 watching
4 forks

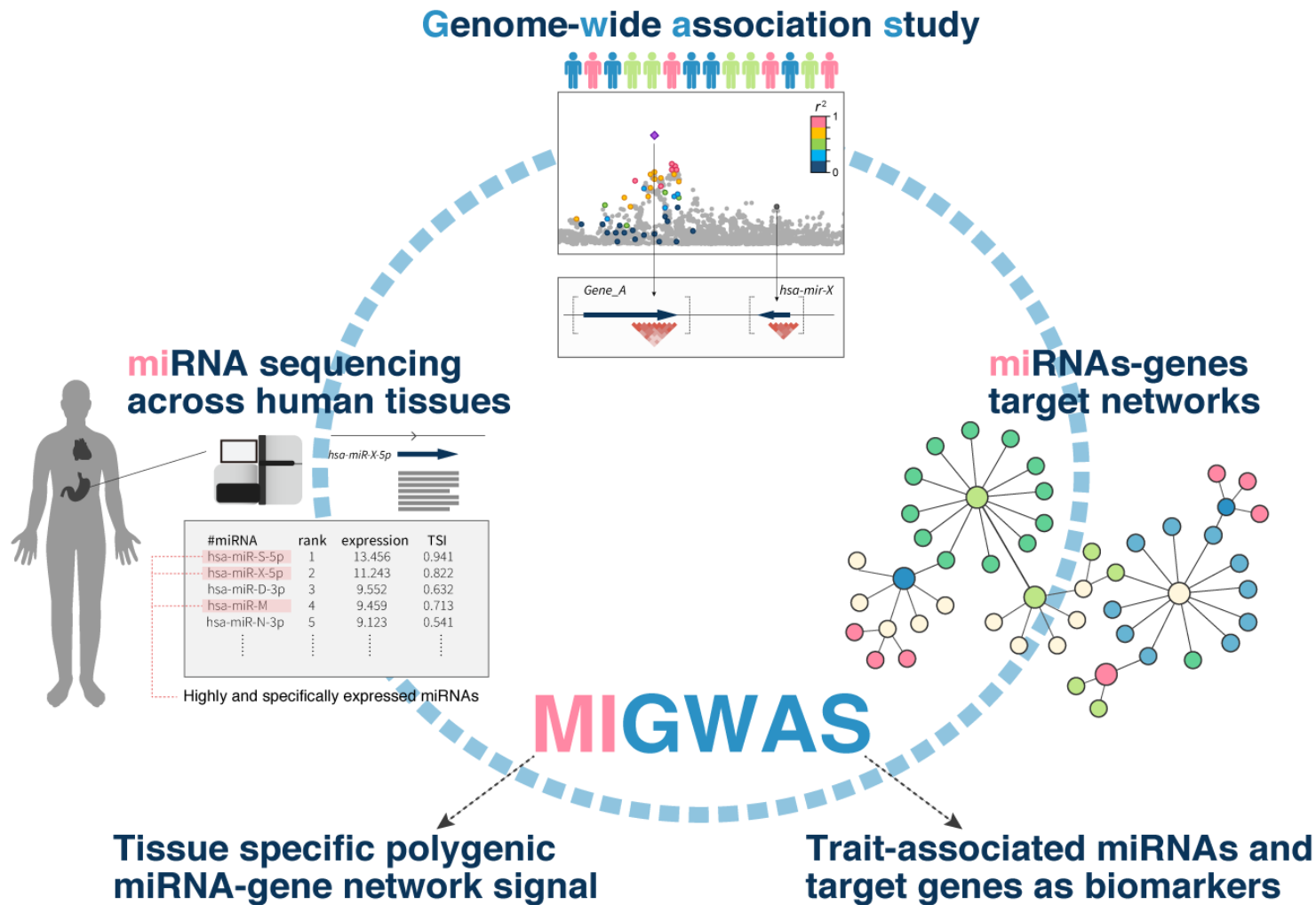
Releases
No releases published

Packages
No packages published

<https://github.com/mkanai/grimon>

•github上でソースコードが公開されています。

④-8: MIGWAS (miRNA-target gene networks enrichment on GWAS)



<https://github.com/saorisakaue/MIGWAS>

- GWAS結果からバイオマーカーmiRNAを同定するMIGWASのサイトです。
- github上でソースコードが公開されています。

Webツール入門

- ① ゲノム・遺伝子情報のWebツール
- ② 遺伝子変異・SNP情報のWebツール
- ③ 疾患感受性遺伝子情報・解析結果のWebツール
- ④ エピゲノム情報のWebツール
- ⑤ 創薬情報のWebツール

⑤-1: DRUGBANK

DRUGBANK Online Browse COVID-19 Search Interaction Checker Downloads Solutions About

Building the foundation for better health outcomes

Access the right information at the right time, with our intelligent clinical drug data API and in-depth knowledge database.

[Learn about our solutions >](#)

Search over 500,000 drugs & drug products on DrugBank Online

Tylenol

Acetaminophen

| | |
|----------------------|--|
| Identifiers | Acetaminophen is an over-the-counter drug used alone or in combination with opioids for pain management, and also as an antipyretic agent. |
| Pharmacology | |
| Indications | |
| Products | |
| Interactions | |
| Chemical Identifiers | |
| References | |
| Chemical Data | |

<http://www.drugbank.ca/>

- 治療薬とその標的遺伝子に関するデータベースサイトです。
- ”Abatacept”と入力してみましょう。

⑤-1: DRUGBANK

DRUGBANK Online

Browse ▾

COVID-19 ▾

Search ▾

Interaction Checker

Downloads

Solutions ▾

About ▾

Drugs ▾



Abatacept

Identification

Pharmacology

Interactions

Products

Categories

Chemical Identifiers

References

Clinical Trials

Pharmacoeconomics

Properties

Targets (2)

Summary Abatacept is a disease-modifying antirheumatic drug (DMARD) used for the management of moderate-to-severe active rheumatoid arthritis and active polyarticular juvenile idiopathic arthritis as monotherapy or in combination with other DMARDs.

Brand Names *Orencia*

| | | | |
|---------------------|-----------|----------------------------------|---------|
| Generic Name | Abatacept | DrugBank Accession Number | DB01281 |
|---------------------|-----------|----------------------------------|---------|

Background Abatacept is a soluble fusion protein, which links the extracellular domain of human cytotoxic T-lymphocyte-associated antigen 4 (CTLA-4) to the modified Fc (hinge, CH2, and CH3 domains) portion of human immunoglobulin G1 (IgG1). Structurally, abatacept is a glycosylated fusion protein with a MALDI-MS molecular weight of 92,300 Da and it is a homodimer of two homologous polypeptide chains of 357 amino acids each. It is produced through recombinant DNA technology in mammalian CHO cells. The drug has activity as a selective co-stimulation modulator with inhibitory activity on T lymphocytes. Although approved for the treatment of rheumatoid arthritis, Repligen has entered a slightly different formulation of CTLA4-Ig into clinical trials (RG2077).

<http://www.drugbank.ca/>

・生物学的製剤(抗体薬)“Abatacept”の性状、標的遺伝子、対象疾患、臨床試験情報、などが提供されます。

⑤-2: TTD (Therapeutic Targets Database)



Therapeutic Target Database



BIDD
Bioinformatics and
Drug Design group

Home Advanced Search ▾ Target Group ▾ Drug Group ▾ Patient Data ▾ Model & Study ▾ Download ▾

Search Whole Database

Search for Targets: ?

Search

Reset

Examples: EGFR; Vascular endothelial growth factor; Peramivir; Renal cell carcinoma ...

Therapeutic Target & Drug Data for Coronavirus (COVID-19, MERS-CoV, SARS-CoV)

A comprehensive collection of anti-coronavirus drugs (small molecular drugs, monoclonal antibodies, protein/peptide drugs, combination drugs, vaccines, etc.) together with their corresponding therapeutic targets data from previous and recent coronavirus researches [\[Click to Explore All Data\]](#)



Jump to the
[Star Target](#) & [Star Drug](#)
for COVID-19 of This
Week

Search for Drugs: ?

Search

Reset

Examples: Cabozantinib; Ebola virus infection; Oseltamivir ...

<http://db.idrblab.net/ttd/>

- 治療薬とその標的遺伝子に関するデータベースサイトです。
- ”Abatacept”と入力してみましょう。

⑤-2: TTD (Therapeutic Targets Database)



Therapeutic Target Database



BIDD
Bioinformatics and
Drug Design group

Home Advanced Search ▾ Target Group ▾ Drug Group ▾ Patient Data ▾ Model & Study ▾ Download ▾

Search for targets

You are searching for: **Abatacept**

| | | | |
|------------------------------|-------------|---|------------------|
| T50912 Target Info | Target Name | T-lymphocyte activation antigen CD86 (FUN1) | |
| | Target type | Successful Target | |
| | Disease | Rheumatoid arthritis; Autoimmune diabetes; | |
| | Drugs | Abatacept | Drug Info |
| T58238 Target Info | Target Name | Activation B7-1 antigen (CD80) | |
| | Target type | Successful Target | |
| | Disease | Rheumatoid arthritis; Non-small-cell lung cancer; | |
| | Drugs | Abatacept | Drug Info |

If You Find Any Error in Data or Bug in Web Service, Please Kindly Report It to **Dr. Wang** and **Dr. Li**.

<https://db.idrblab.net/ttd/>

・生物学的製剤(抗体薬)“Abatacept”の性状、標的遺伝子、対象疾患、臨床試験情報、などが提供されます。

⑤-3: Connectivity Map

CLUE

Tools Projects Developer Help | [Log in](#)

ConnectivityMap

Unravel biology with the world's largest perturbation-driven gene expression dataset.

Start exploring the data by using the text-box on this page to look up perturbagens of interest in Touchstone. To see the suite of tools, including apps to query your gene expression signatures and analyze resulting connections, click on Tools in the menu bar.

> TYPE COMPOUND, GENE, MoA, OR PERTURBAGEN CLASS TO SEE OVERVIEW
> TYPE A SLASH CHARACTER "/" TO SEE LIST OF COMMANDS

DATA VERSION: Beta / SOFTWARE VERSION: 1.1.1.43

SUPPORT + OFFICE HOURS CANCELED

As of 7/15/22, our team will no longer be offering office hours and email support for clue.io and its app. Our extensive online learning resources will still be available through [Connectopedia](#), including the comprehensive CLUE glossary. Please note that this will not affect any functionalities on clue.io and its apps—we are only ending individualized support. As always, we appreciate your continued use of clue.io! —The CLUE Team .

Data and Tools

We are excited to announce the release of the updated CMap LINCS gene expression resource. This release is an expansion upon the previous 2017 data release and contains >3M gene expression profiles and >1M replicate-collapsed signatures.

1. These data are available for download from the [LINCS data releases app](#) as well as the from the [clue data library](#).
2. The data can be queried with external gene sets using the [clue query app](#).
3. We also provide a web application for [querying the metadata](#).
4. And a python library for accessing the [data programmatically](#).

In addition, we provide the following tools to help facilitate data access and use:

1. Code libraries for accessing and analyzing [CMap data](#)
2. Notebooks that illustrate common modes of [data access and analysis](#)
3. Docker containers for running common [analysis algorithms](#)

Please note that these data and tools are released as a

<https://clue.io/about>

• 米国Broad研究所が提供する、化合物投与時の遺伝子発現量や蛋白
量の変化を対象としたデータベースです。

⑤-4: Anatomical Therapeutic Chemical (ATC) Classification

KEGG Anatomical Therapeutic Chemical (ATC) Classification

[[Brite menu](#) | [Download htext](#) | [Download json](#) | [Help](#)]

Search

▼ ▼ ▼ ▼ ▼ ▼ One-click mode

▶ A ALIMENTARY TRACT AND METABOLISM

▶ B BLOOD AND BLOOD FORMING ORGANS

▶ C CARDIOVASCULAR SYSTEM

▶ D DERMATOLOGICALS

▶ G GENITO URINARY SYSTEM AND SEX HORMONES

▶ H SYSTEMIC HORMONAL PREPARATIONS, EXCL. SEX HORMONES AND INSULINS

▶ J ANTIINFECTIVES FOR SYSTEMIC USE

▶ L ANTINEOPLASTIC AND IMMUNOMODULATING AGENTS

▶ M MUSCULO-SKELETAL SYSTEM

▶ N NERVOUS SYSTEM

▶ P ANTIPARASITIC PRODUCTS, INSECTICIDES AND REPELLENTS

▶ R RESPIRATORY SYSTEM

▶ S SENSORY ORGANS

▶ V VARIOUS

[[DRUG](#) | [BRITE](#) | [KEGG2](#) | [KEGG](#)]

Last updated: July 21, 2022

According to WHO's ATC 2022

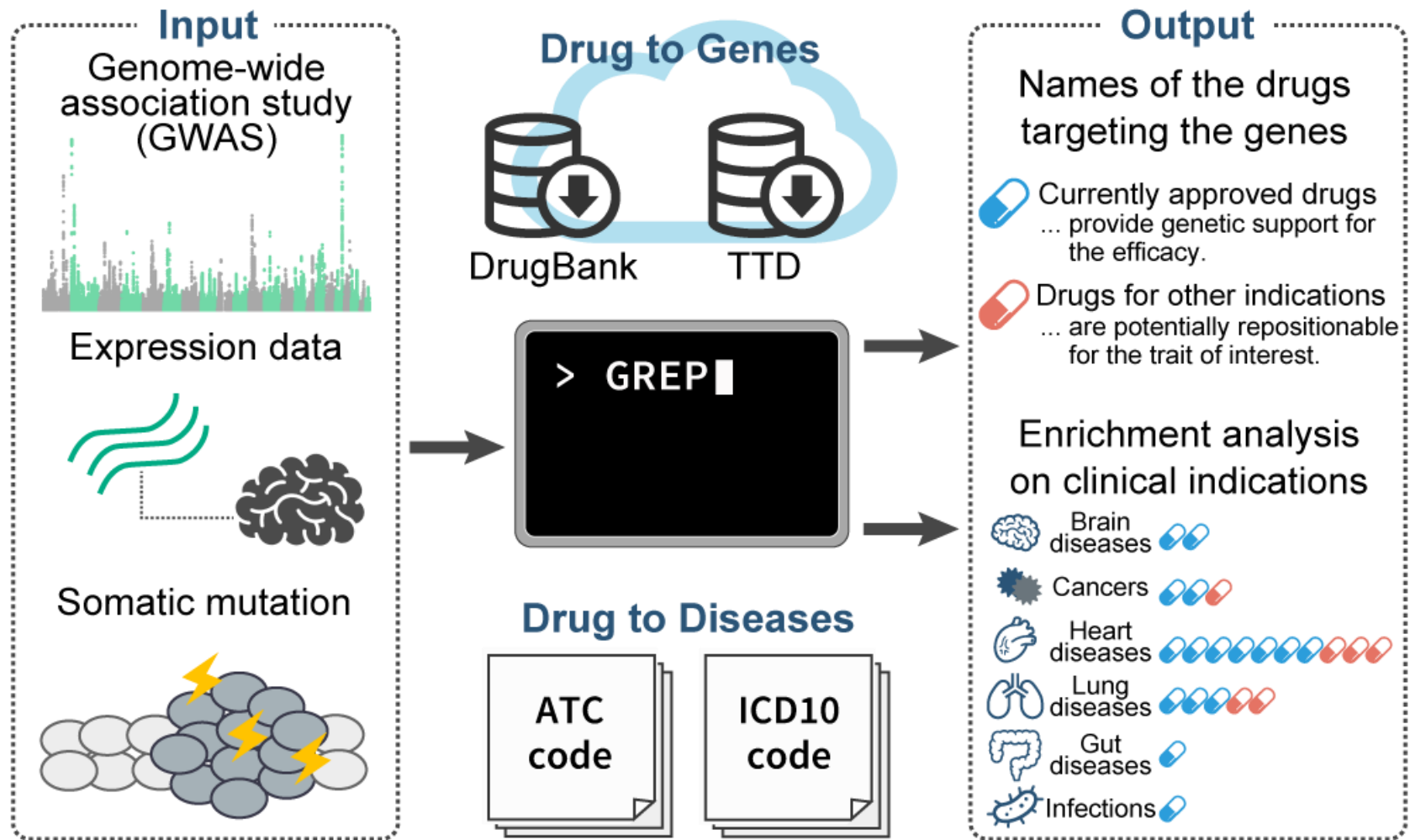
» [Japanese version](#)

http://www.kegg.jp/kegg-bin/get_htext?br08303.keg

•WTOのATC分類に基づく、疾患と治療薬の網羅的な対応表です。

(Kyoto Encyclopedia of Genes and Genomes(KEGG)上のページになります。)⁷²

⑤-5: GREP (Genome for REPositioning drugs)



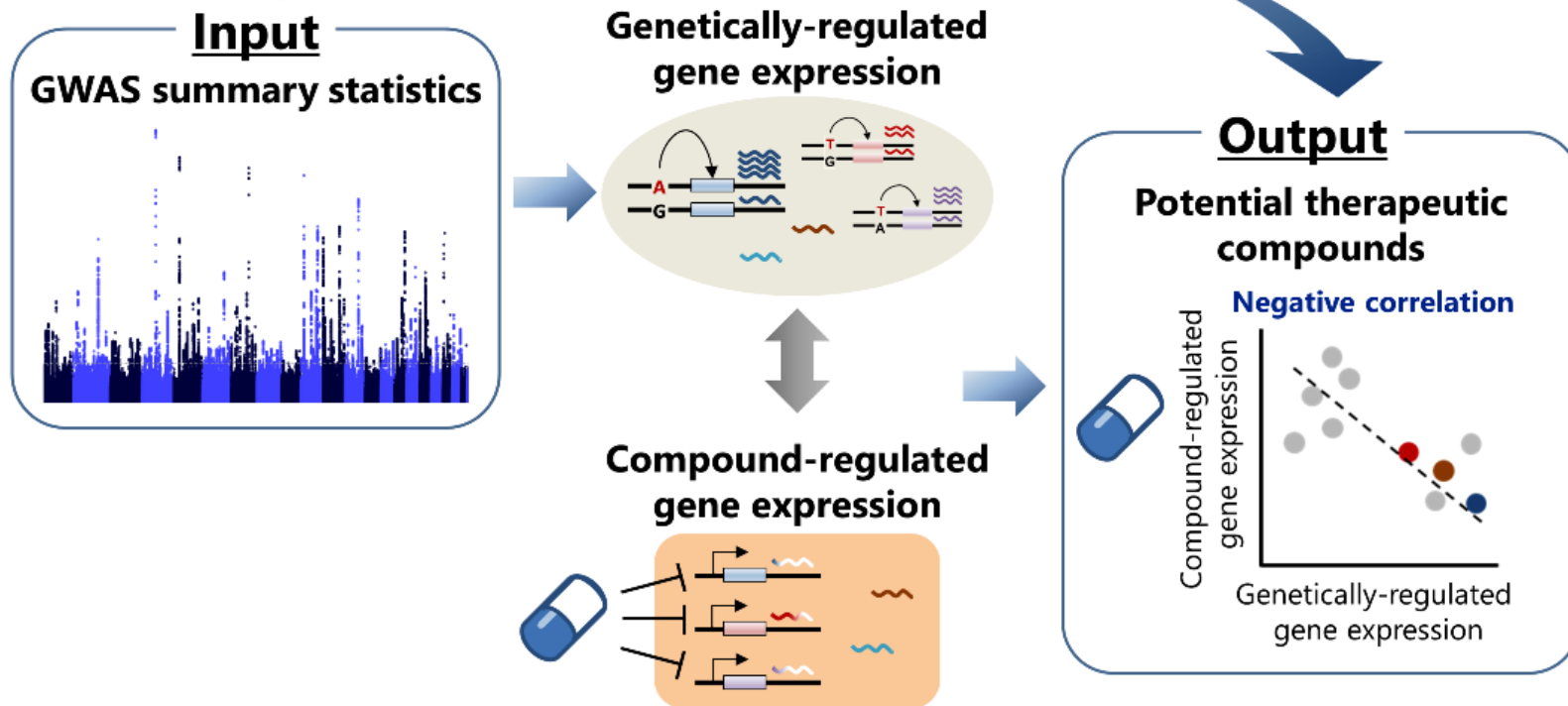
<https://github.com/saorisakaue/GREP>

• ATC分類や創薬データベースに基づき、疾患関連遺伝子からリポジショニング候補治療薬や適応候補疾患をスクリーニングするツールです。⁷³

⑤-6: Trans-Phar (integration of TWAS and pharmacological database)

Trans-Phar

Integration of **Trans**criptome-wide association study and **Phar**macological database



<https://github.com/konumat/Trans-Phar>

• TWAS (Transcriptome-Wide Association Study) を用いて、GWAS 結果から遺伝子発現量変化を推定し、化合物をスクリーニングする創薬ツール⁷⁴です。

⑤-7:STITCH

Version: 5.0

LOGIN | REGISTER

STITCH

Search Download Help My Data

Welcome to STITCH

Chemical-Protein Interaction Networks

| ORGANISMS | CHEMICALS | PROTEINS | INTERACTIONS |
|-----------|-----------|----------|--------------|
| 2031 | 0.5 mio | 9.6 mio | 1.6 bn |

SEARCH

<http://stitch.embl.de>

- 化合物とタンパク質が構成するネットワーク情報のサイトです。
- ”PADI2”と入力してみましょう。

⑤-7:STITCH

Version: 5.0

LOGIN | REGISTER

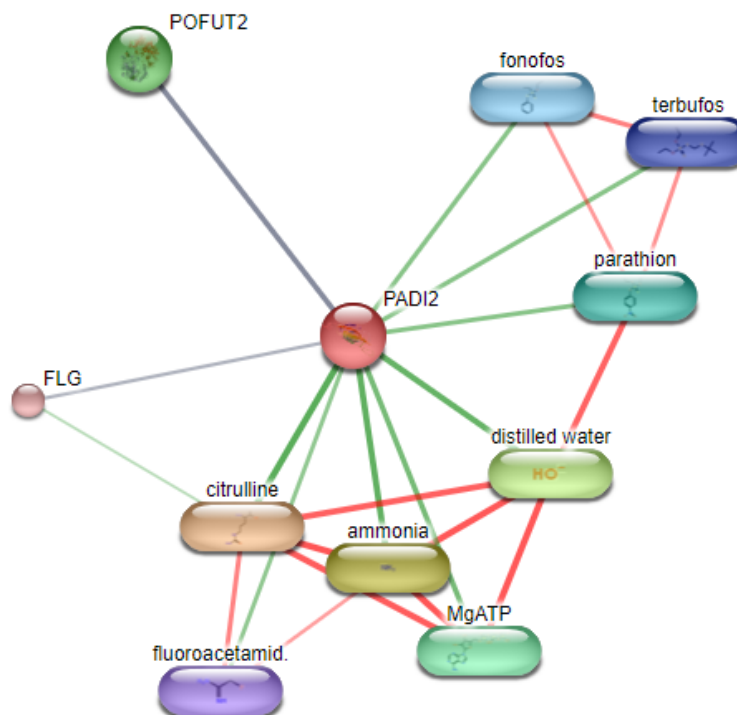
STITCH

Search

Download

Help

My Data



This is the **confidence view**. Stronger associations are represented by thicker lines.

Protein-protein interactions are shown in grey, chemical-protein interactions in green and interactions between chemicals in red.

<http://stitch.embl.de>

- PADI2遺伝子(タンパク質)と相互作用をもつ化合物や、他のタンパク質とのネットワーク情報が、図として提供されます。

⑤-8: STRING

Version: 11.5

LOGIN REGISTER SURVEY



Search Download Help My Data

Welcome to STRING

Protein-Protein Interaction Networks
Functional Enrichment Analysis

ORGANISMS
14094

PROTEINS
67.6 mio

INTERACTIONS
>20 bln

SEARCH

<http://string-db.org/>

- タンパク質間相互作用ネットワーク情報のサイトです。
- ”PADI2”と入力してみましょう。

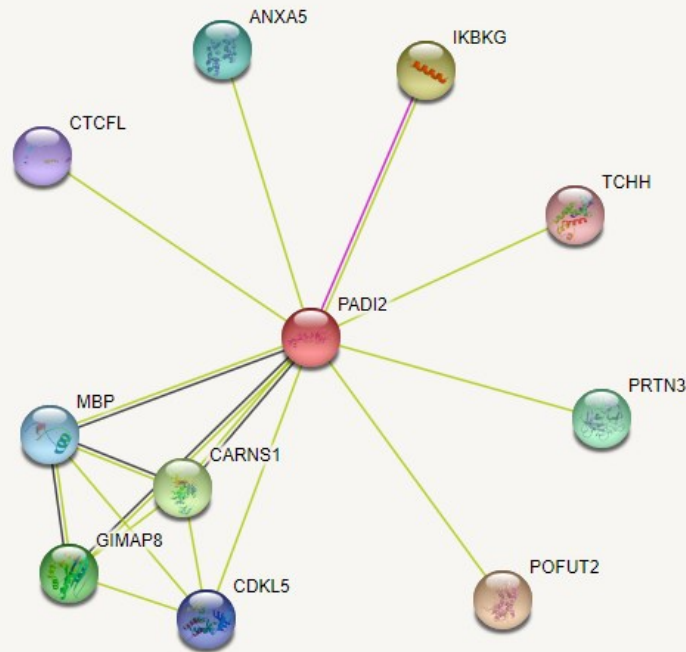
⑤-8:STRING

Version: 11.5

LOGIN REGISTER SURVEY



Search Download Help My Data



Viewers > Legend > Settings > Analysis > Exports > Clusters > More > Less >

Nodes:

Network nodes represent proteins

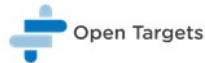
Node Color

Node Content

<http://string-db.org/>

•PADI2遺伝子(タンパク質)が相互作用するタンパク質とのネットワーク情報が、図として提供されます。

⑤-9: Open Targets



About ▾ Research ▾ Jobs Blog Community Contact



Developing safe and effective drugs is difficult and expensive

We are dedicated to changing this with innovative experimental and informatics approaches

Open Targets is an innovative, large-scale, multi-year, public-private partnership that uses human genetics and genomics data for systematic drug target identification and prioritisation.

Visit the [Open Targets Platform](#) which integrates public domain data to enable target identification and prioritisation, or the [Open Targets Genetics portal](#) which identifies targets based on GWAS and functional genomics. We complement data integration with large scale systematic experimental approaches to support target identification, prioritisation and validation. Check out [our latest papers](#) describing our experimental target identification approaches in oncology, neurodegeneration, and immunity and inflammation.

Find targets for a given disease

[Visit the Open Targets Platform](#)

Discover genetic evidence for targets

[Visit Open Targets Genetics](#)

Explore our experimental projects

[Read our research publications](#)

<https://www.opentargets.org/>

・ゲノム創薬解析を念頭に、創薬ターゲットの情報をまとめたデータベースです。

終わりに

- **ゲノム研究や遺伝統計解析を実施する際に便利な、Webツールを挙げてみました。**
- **今回は、Webブラウザ上の簡単な操作で情報が得られるツールを対象に紹介してみました。**
- **こんなのあったら便利だな、というツールは、だいたい実装されています。**
- **Webツールを使いこなすコツは、“とりあえず触ってみる”ことです。**
- **詳しい機能を覚えるのは後回しにして、“どんなことができるWebツールが世の中にあるのか”を体感してもらえればと思います。**