

多型データベースの紹介 (TogoVarなど)

令和元年8月7日 @AJACS番町3

国立研究開発法人 科学技術振興機構 (JST)
バイオサイエンスデータベースセンター (NBDC)
三橋 信孝

多型/バリアントとは

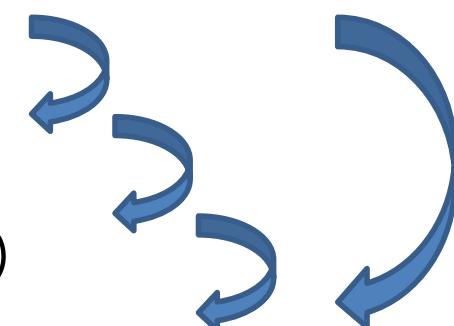
- 多型(polymorphism) / バリアント (variant)
 - ヒトゲノムにおいて観察されるreferenceと異なる塩基配列のこと
 - 多型:集団内での頻度1%以上
- ゲノムと形質との関連を明らかにするうえで重要な基礎データ

ゲノム、エピゲノム

トランск립トーム

プロテオーム

形質(疾患、薬剤副反応、形態等)



多型/バリアントの種類

Types of genetic variation

Genetic variation is commonly divided into three main forms:

Single base-pair substitution

There are also known as single nucleotide polymorphisms (SNPs) and can be any nucleic acid substitution:

- Transition
 - interchange of the purine (Adénine/Guanine)
 - or pyrimidine (Cytosine/Thymine) nucleic acids
- Transversion
 - interchange of a purine and pyrimidine nucleic acid (Figure 3)

Move the slider to the right to see a SNP :

DNA with SNP

ACTGACGCATGCATCAT**T**CATGC

Figure 3 SNPs result from the substitution of a single base-pair. In this example we have a transversion event substituting a Thymine nucleic acid in place of a Guanine.

Insertion or deletion, also known as 'indel'

Insertion or deletion of a single stretch of DNA sequence that can range from two to hundreds of base-pairs in length (Figure 4).

Reference	ACTGACGCATGCATCATGCATGC
Insertion	ACTGACGCATGG T A CATCATGCATGC
Deletion	ACTGACG--TGCATCATGCATGC

Figure 4 Indels affect a string of base-pairs. In this example the insertion string shows that GTA has been inserted, and the deletion string shows a deletion of CA.

一塩基多型

(SNP: Single Nucleotide Polymorphism)
(個人当たり～400万)

18番目のGがTに置換された

挿入(insertion)・欠失(deletion)
(個人当たり～70万)
(長さ～数百bp)

<https://www.ebi.ac.uk/training/online/course/human-genetic-variation-i-introduction-2019/what-genetic-variation/types-genetic-variation>

多型/バリエントの種類

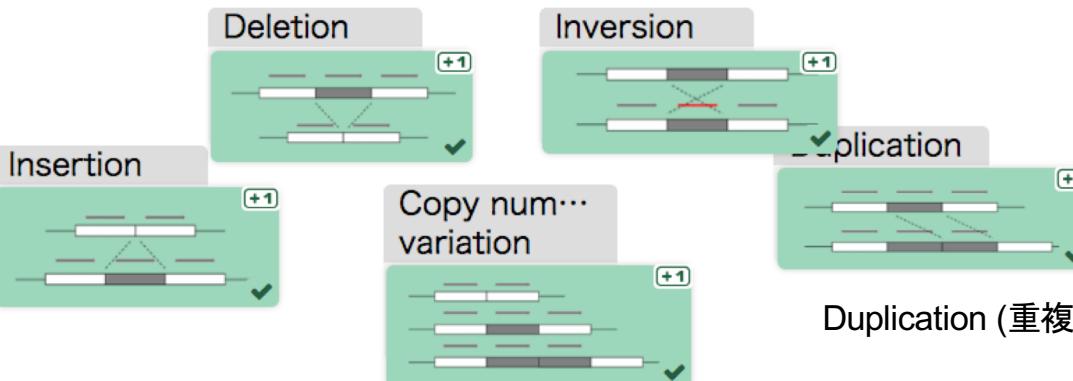
構造多型 (SV: Structural Variant) : 長いものを指す(50bp～)
(個人当たり 1～2万)

Structural variation

Typically used to describe genetic variation that occurs over a larger DNA sequence. This category of genetic variation includes both copy number variation and chromosomal rearrangement events. Discover the five most common types of structural variants in this drag-and-drop game:

Drag and drop the images into the correctly labelled area

Inversion (逆位)



欠失と重複をCopy Number Variation (CNV)ともいう

★ 5 / 5

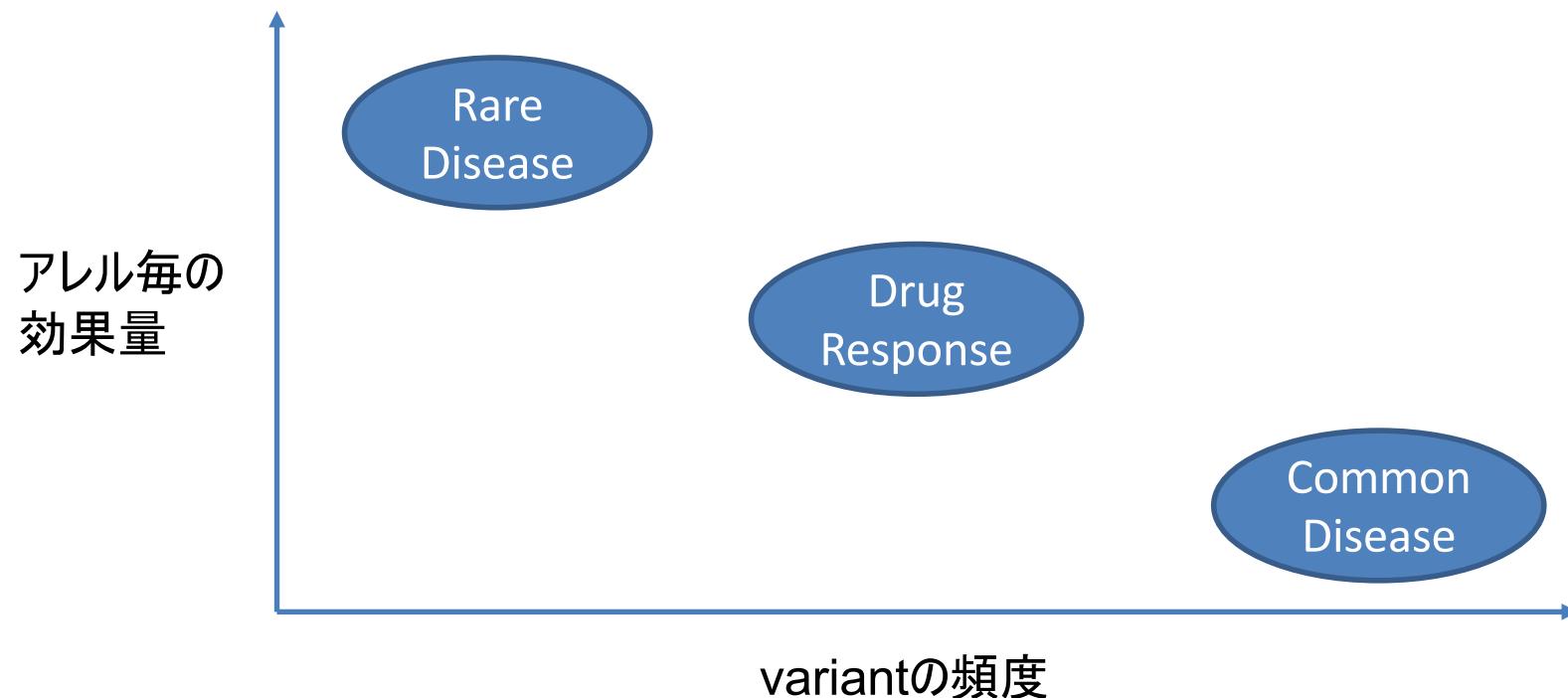
今回の対象外

<https://www.ebi.ac.uk/training/online/course/human-genetic-variation-i-introduction-2019/what-genetic-variation/types-genetic-variation>

4

バリエントの頻度と形質の関係

- Common Disease Common Variant仮説
- Rare Disease Rare Variant仮説

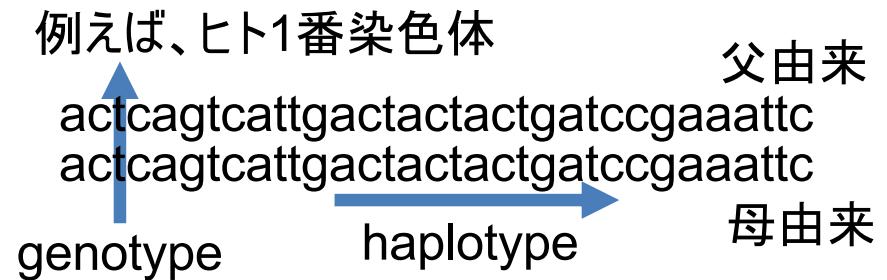


疾患と関連する遺伝子同定の研究概要

- ありふれた疾患 (common disease)
Rare VariantはPrimaryの探索から外されることが多い。
 - 稀少疾患 (rare disease)
Common VariantはPrimaryの探索から外されることが多い。
 - Trio解析
 - 発症者 + 未発症の父母の3人で、発症者におけるde novoな variantを抽出
- 一般集団のvariant頻度情報は基礎的情報として有用

進化するプラットフォーム

- ・サンガーシーケンサー
 - 配列情報
- ・SNP Chip
 - 60-90万箇所のgenotype情報
- ・次世代(第2世代)シーケンサー(例:Illumina)
 - リード長: 数十bから数百b
 - 全exome領域や全genome領域等のgenotype情報、haplotype情報
- ・第3世代シーケンサー(例:PacBio)
 - リード長: 数kbから数十kb
 - 特定の領域のgenotype情報、haplotype情報、構造多型の検出

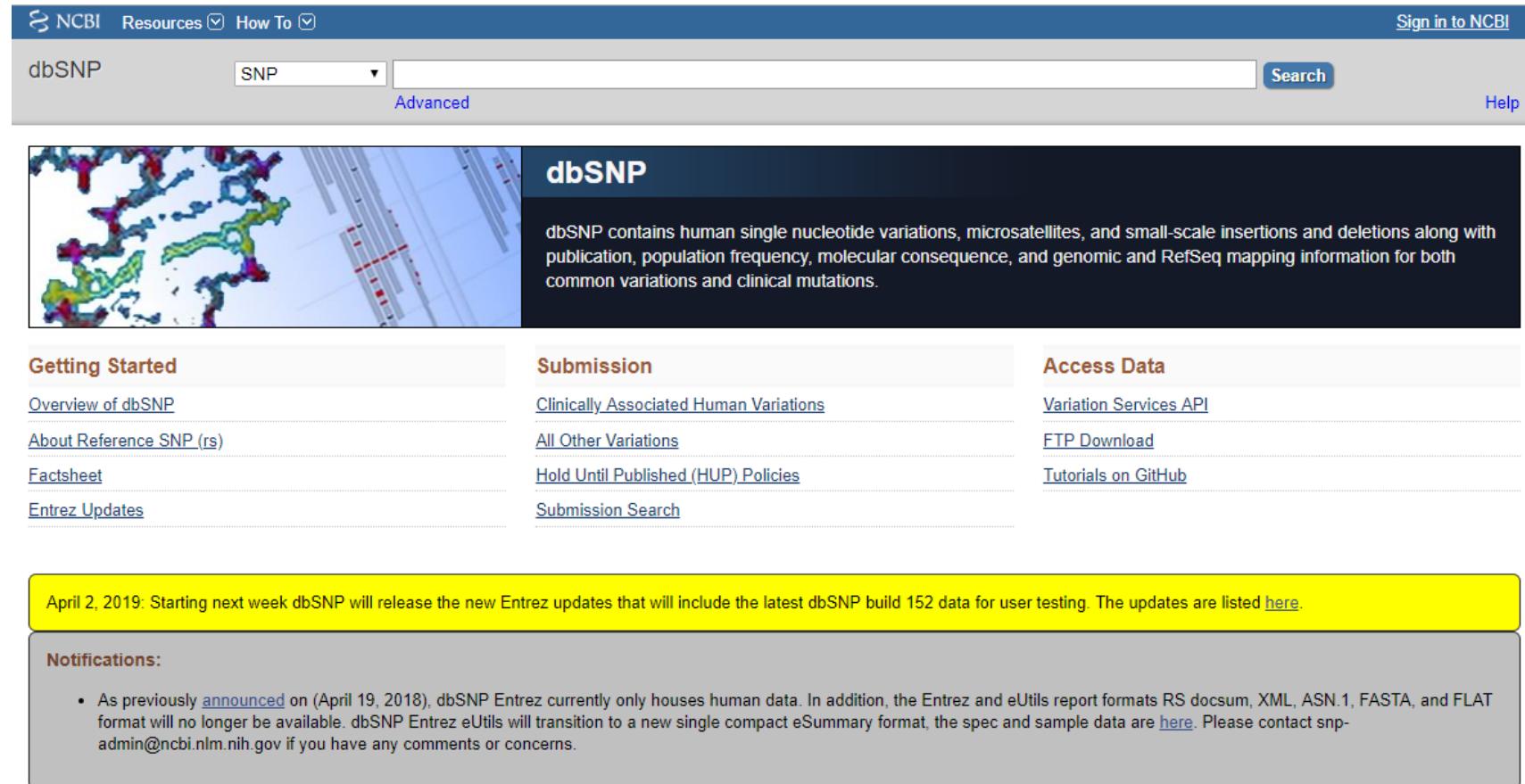


多型データベースの紹介 1:dbSNP

dbSNP

- 運営元：米国NCBI（National Center for Biotechnology Information）
- URL: <https://www.ncbi.nlm.nih.gov/snp/>
- 特徴
 - 様々の研究において検知された多型/バリアントの情報を受け付け、ss番号を付与。その後、ss番号をまとめてrs(refSNP)番号を付与
 - ss番号 → rs番号
 - 50bp以下の多型を対象に情報を収集
 - 50bpよりも大きい多型 dbVarに収載
 - 多型の有無の確認
 - 6億バリアント

dbSNPの画面説明、操作



The screenshot shows the dbSNP homepage. At the top, there's a navigation bar with links for NCBI, Resources, How To, Sign in to NCBI, and Help. Below the navigation is a search bar with dropdown menus for "dbSNP" and "SNP", and a "Search" button. There's also an "Advanced" link. The main content area features a large image of a genome map on the left and a dark panel on the right with the title "dbSNP". The panel describes dbSNP as containing 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. Below this are three sections: "Getting Started" (Overview of dbSNP, About Reference SNP (rs), Factsheet, Entrez Updates), "Submission" (Clinically Associated Human Variations, All Other Variations, Hold Until Published (HUP) Policies, Submission Search), and "Access Data" (Variation Services API, FTP Download, Tutorials on GitHub). A yellow banner at the bottom left announces: "April 2, 2019: Starting next week dbSNP will release the new Entrez updates that will include the latest dbSNP build 152 data for user testing. The updates are listed [here](#)". A "Notifications:" section below the banner contains a bullet point about changes to the Entrez and eUtils report formats.

Notifications:

- As previously announced on (April 19, 2018), dbSNP Entrez currently only houses human data. In addition, the Entrez and eUtils report formats RS docsum, XML, ASN.1, FASTA, and FLAT format will no longer be available. dbSNP Entrez eUtils will transition to a new single compact eSummary format, the spec and sample data are [here](#). Please contact snp-admin@ncbi.nlm.nih.gov if you have any comments or concerns.

dbSNP News and Announcements

[RSS Feed](#)  dbSNP News and Announcements(RSS) Feed

[Email List](#)

Related Sites

[Variation Portal](#)

[Variation Tools](#)

Variation Databases

[dbVar](#)

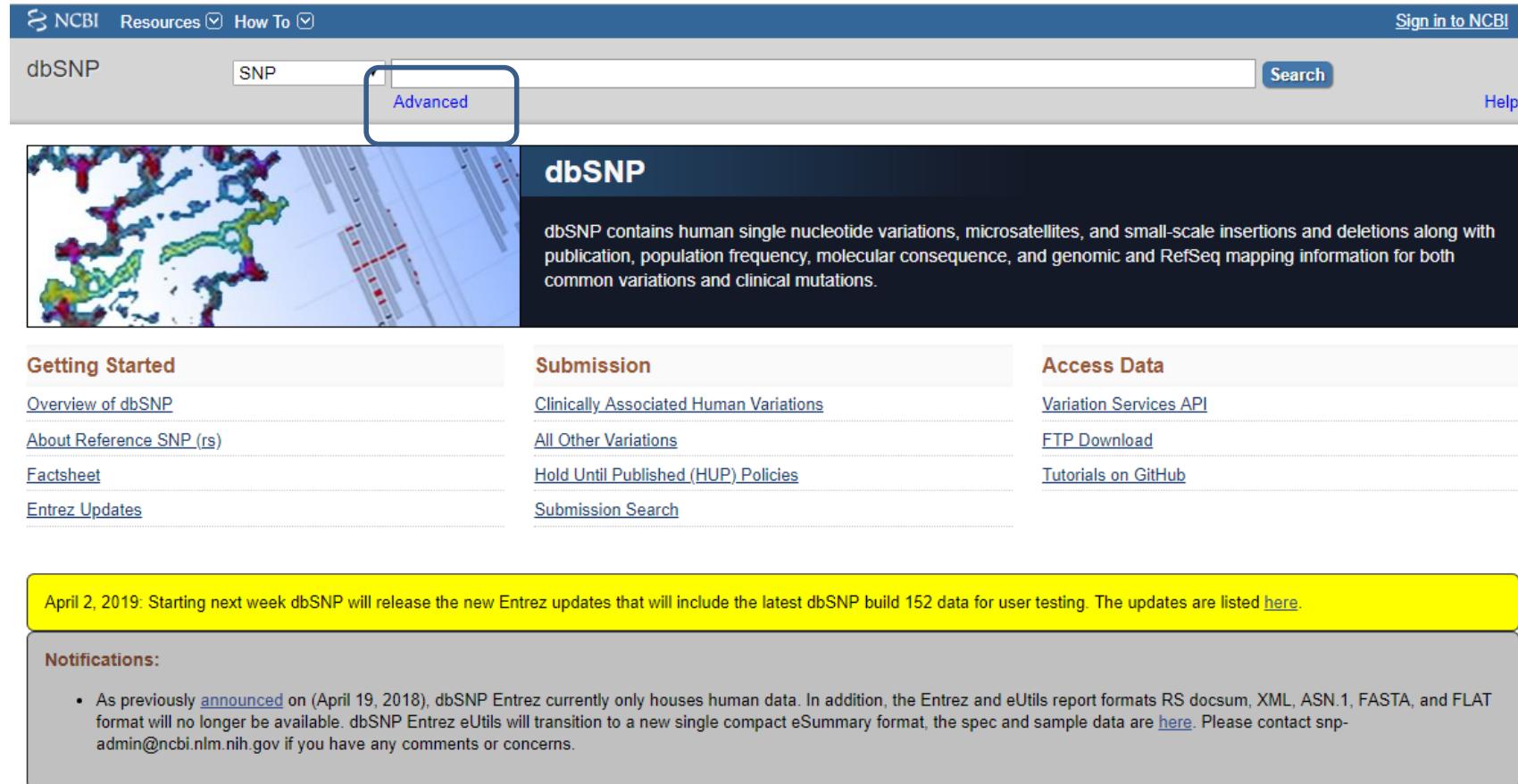
[dbGaP](#)

[ClinVar](#)

[GTR](#)

検索対象語を入力だけ→何を対象に調べているのか？

dbSNPにおける拡張検索機能



The screenshot shows the dbSNP homepage. At the top, there is a navigation bar with links for NCBI, Resources, How To, Sign in to NCBI, and Help. Below the navigation bar, the dbSNP logo is on the left, followed by tabs for SNP and Advanced (which is highlighted with a blue box). A search bar and a "Search" button are to the right of the tabs. The main content area features a map-like visualization on the left and a dark panel on the right containing the dbSNP logo and a brief description of the database's content. Below this, there are three columns: "Getting Started" (Overview of dbSNP, About Reference SNP (rs), Factsheet, Entrez Updates), "Submission" (Clinically Associated Human Variations, All Other Variations, Hold Until Published (HUP) Policies, Submission Search), and "Access Data" (Variation Services API, FTP Download, Tutorials on GitHub). A yellow banner at the bottom of the main content area contains a message about upcoming Entrez updates. A "Notifications:" section below the banner contains a bullet point about changes to dbSNP Entrez data formats.

April 2, 2019: Starting next week dbSNP will release the new Entrez updates that will include the latest dbSNP build 152 data for user testing. The updates are listed [here](#).

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dbSNP News and Announcements

[RSS Feed](#)  [dbSNP News and Announcements\(RSS\) Feed](#)

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Related Sites

[Variation Portal](#)

[Variation Tools](#)

Variation Databases

[dbVar](#)

[dbGaP](#)

[ClinVar](#)

[GTR](#)

dbSNPの拡張検索の組み立て

NCBI Resources How To Sign in

SNP Home Help

SNP Advanced Search Builder

Use the builder below to create your search

Edit Clear

Builder

All Fields		Show index list
AND	Accession	Show index list
Search	All Fields	Show index list
	Allele	
	Base Position	
History	Chromosome	Download history Clear history
Search	Clinical Significance	
#4	Contig Position	
#3	Create Build ID	
#2	Filter	
#1	Function Class	
	Gene Description	
	Gene Name	

You are here: NCBI > Variation >

GETTING STARTED

- NCBI Education
- NCBI Help Manual
- NCBI Handbook
- Training & Tutorials
- Submit Data

Genotype

Global Minor Allele Frequency

Local SNP ID

LocusLink ID

Method Class

Modification Date

Organism

POPULAR

- PubMed
- Bookshelf
- PubMed Central
- BLAST
- Nucleotide
- Genome
- SNP
- Gene
- Protein
- PubChem

FEATURED

- Genetic Testing Registry
- GenBank
- Reference Sequences
- Gene Expression Omnibus
- Genome Data Viewer
- Human Genome
- Mouse Genome
- Influenza Virus
- Primer-BLAST
- Sequence Read Archive

NCBI INFORMATION

- About NCBI
- Research at NCBI
- NCBI News & Blog
- NCBI FTP Site
- NCBI on Facebook
- NCBI on Twitter
- NCBI on YouTube
- Privacy Policy

Advanced
Searchを使用
しないと、
検索語を
All Fields
に対して検索

dbSNPの検索結果 (PDCD1 rs11568821)

NCBI Resources How To Sign in to NCBI

dbSNP SNP PDCD1 rs11568821 Search Create alert Advanced Help

Annotation Cited in PubMed PubMed nucleotide Global MAF Custom range... Validation Status by-cluster by-frequency Clear all Show additional filters

Display Settings: Summary, Sorted by SNP_ID Send to: Filters: Manage Filters Find related data Database: Select

検索式の表示

Find items

Search details PDCD1 [All Fields] AND rs11568821 [All Fields]

Search See more...

過去の検索内容の表示

Recent activity Turn Off Clear

- PDCD1 rs11568821 (2) SNP
- PDCD1 (3020) SNP
- 12 : 111803962 (5) SNP
- rs671 (5) SNP

See more...

検索候補が表示

rs11568821 [Homo sapiens]
 1. Variant type: SNV
 Alleles: C>G,T
 Chromosome: NT_187527.1:63858
 Gene: LOC105373977 (GeneView), PDCD1 (GeneView)
 Functional Consequence: downstream_transcript_variant,genic_downstream_transcript_variant,upstream_transcript_variant,intron_variant
 Clinical significance: risk-factor
 Validated: by frequency,by cluster
 Global MAF: T=0.0409/205 (1000Genomes)
 T=0.0711/2196 (GnomAD)
 T=0.0746/9373 (TOPMED)
 T=0.1157/446 (ALSPAC)
 T=0.1162/431 (TWINSUK)
 HGVS: NC_000002.12:g.241851760C>G, NC_000002.12:g.241851760C>T,
 NC_000002.11:g.242793912C>G, NC_000002.11:g.242793912C>T,
 NG_012110.1:g.12147G>C, NG_012110.1:g.12147G>A, NT_187527.1:g.63858C>G,
 NT_187527.1:g.63858C>T
 PubMed Varview

rs606231173 has merged into rs11568821 [Homo sapiens]
 2. Variant type: SNV
 Alleles: C>G,T
 Chromosome: NT_187527.1:63858
 Gene: LOC105373977 (GeneView), PDCD1 (GeneView)

dbSNPの検索結果(詳細画面)

NIH U.S. National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP Short Genetic Variations

Search for rs Example: rs268

Search

旧表示形式
での表示も可能

Reference SNP (rs) Report

← Switch to classic site

rs11568821

Organism: *Homo sapiens*

Position: chr2:241851760 (GRCh38.p12) ⓘ

Alleles: C>G / C>T

Variation Type: SNV Single Nucleotide Variation

Frequency: T=0.07464 (9373/125568, TOPMED)
T=0.0711 (2196/30874, GnomAD)
T=0.041 (205/5008, 1000G) (+ 2 more)

Clinical Significance: Reported in ClinVar

Gene : Consequence: PDCD1 .intron Variant
LOC105373977 : 2KB Upstream Variant

Publications: 32 citations

Genomic View: See rs on genome

Current Build 152
Released October 2, 2018

ClinVarへのリンク

Variation Viewer

Variant Details

- Clinical Significance
- Frequency
- Aliases
- Submissions
- History
- Publications

Genomic Placements

Sequence name	Change
GRCh37.p13 chr 2	NC_000002.11:g.242793912C>T
GRCh37.p13 chr 2	NC_000002.11:g.242793912C>G
GRCh38.p12 chr 2	NC_000002.12:g.241851760C>T
GRCh38.p12 chr 2	NC_000002.12:g.241851760C>G
GRCh38.p12 chr 2 alt locus HSCHR2_3_CTG15	NT_187527.1:g.63858C>T
GRCh38.p12 chr 2 alt locus HSCHR2_3_CTG15	NT_187527.1:g.63858C>G
PDCD1 RefSeqGene	NG_012110.1:g.12147G>A
PDCD1 RefSeqGene	NG_012110.1:g.12147G>C

参照配列毎の位置
ClinVarの情報
頻度情報
Submission情報
rs番号の変遷
文献情報
をTabで切り替えられます

dbSNPの旧表示形式

NCBI dbSNP Short Genetic Variations

Search small variations in dbSNP or large structural variations in dbVar

Search Entrez | dbSNP ▾ for | Go

Reference SNP (refSNP) Cluster Report: rs11568821 ** With other allele **

RefSNP	Allele	HGV Names	Links
Organism: human (<i>Homo sapiens</i>)	SNV: single nucleotide variation	CM000664.2:g.241851760>T NC_000002.11:g.242793912C>G NC_000002.11:g.242793912C>T NC_000002.12:g.241851760>G NC_000002.12:g.241851760C>T NG_012110.1:g.12147G>A NG_012110.1:g.12147G>C NM_005018.2:c.627+189G>A NM_005018.2:c.627+189G>C NT_187527.1:g.63858C>G NT_187527.1:g.63858C>G NT_187527.1:g.63858C>G NT_187527.1:g.63858C>G	...more
Molecule Type: Genomic	RefSNP Alleles: A/C/G (REV)		
Created/Updated in build: 120/151	Allele Origin: C:germline G:germline		
Map to Genome Build: 108/Weight 1	Ancestral Allele: G		
Validation Status:	Variation Viewer: VarView		
Citation: PubMed LitVar NEW	Clinical Significance: With other allele [ClinVar]		
MAF/MinorAlleleCount: T=0.0409/205 (1000 Genomes)			
	T=0.0746/9373 (TOPMED)		

SNP Details are organized in the following sections: GeneView Map Submission Fasta Resource Diversity Validation

Integrated Maps (Hint: click on 'Chr Pos' to see variant in the new NCBI variation viewer)

Assembly	Annotation Release	Chr	Chr Pos	Contig	Contig Pos	SNP to Chr	Contig allele	Contig to Chr	Neighbor SNP	Map Method
GRCh38.p7	108	2	NA	NT_187527.1	63858	NA	C	NA	view	mapup
GRCh38.p7	108	2	241851780	NT_005403.18	147355745	Rev	C	Fwd	view	mapup
GRCh37.p13	105	2		NT_005418.13	1984780	Rev	C	Fwd	view	blast

GeneView via analysis of contig annotation: PDCD1 programmed cell death 1

View more variation on this gene (click to hide).

Clinical Source: in gene region cSNP has frequency double hit [Go](#)

Primary Assembly Mapping

Assembly	SNP to Chr	Chr	Chr position	Contig	Contig position	Allele
GRCh38.p7	Rev	2	241851780	NT_005403.18	147355745	C

RefSeqGene Mapping

RefSeqGene	Gene (ID)	SNP to RefSeqGene	Position	Allele
NG_012110.1	PDCD1 (5133)	Fwd	12147	G

Gene Model(s)

Function	SNP to mRNA	Accession	Position	Allele change	Accession	Position	Residue change
nearGene-3	NA	XM_008712573.2	NA	NA ⇒ NA	NA	NA	NA

NC_000002.12 | Find: | 241,851,710 | 241,851,720 | 241,851,730 | 241,851,740 | 241,851,750 | **rs11568821** | 241,851,770 | 241,851,780

Genes, NCBI Homo sapiens Annotation Release 109, 2018-03-27

Suspect_variations, dbSNP Build 151 (Homo sapiens Annotation Release 108)

dbSNPである遺伝子を対象に検索

NCBI Resources How To

dbSNP SNP PDCD1 Create alert Advanced

Annotation Cited in PubMed PubMed nucleotide protein structure

Display Settings: Summary, 20 per page, Sorted by SNP_ID

Search results Items: 1 to 20 of 3020 << First < Prev Page 1 of 151 Next

Function Class frame shift stop gained

Global MAF Custom range...

Validation Status by-cluster by-frequency

[Clear all](#)

[Show additional filters](#)

rs11568821 [*Homo sapiens*]

1.

Variant type:	SNV
Alleles:	C>G,T
Chromosome:	NT_187527.1:63858
Gene:	LOC105373977 (GeneView), PDCD1 (GeneView)

Functional Consequence: downstream_transcript_variant,genic_downstream_transcript_variant,upstream_transcript_variant,in

Clinical significance: risk-factor

Validated: by frequency,by cluster

Global MAF: T=0.0409/205 (1000Genomes)
T=0.0711/2196 (GnomAD)
T=0.0746/9373 (TOPMED)
T=0.1157/446 (ALSPAC)
T=0.1162/431 (TWINSUK)

HGVS: NC_000002.12:g.241851760C>G, NC_000002.12:g.241851760C>T,
NC_000002.11:g.242793912C>G, NC_000002.11:g.242793912C>T,

dbSNPの遺伝子ビュー

NCBI

dbSNP
Short Genetic Variations

dbVar ClinVar GaP PubMed Nucleotide Protein

Search small variations in dbSNP or large structural variations in dbVar

Search Entrez **dbSNP** for Go

Have a question about dbSNP? Try searching the SNP FAQ Archive! Go

SNP linked to Gene (geneID:5133) Via Contig Annotation

The SNP GeneView page only reports human variation on GRCh38. A new Variation Viewer is available to view the gene PDCD1 variations in GRCh37/p13 or GRCh38, and will replace SNP GeneView later this year. Please visit the [Help Page](#) or [YouTube](#) for available features and send your comments and suggestions to NCBI [helpdesk](#).

Send rs# on all gene models to Batch Query Download all rs# to file.

Gene Model (mRNA alignment) information from genome sequence

Total gene model (contig mRNA transcript):				4		
mRNA	transcript	protein	mRNA orientation	Contig	Contig Label	List SNP
NM_005018.2	minus strand	NP_005009.2	reverse	NT_005403.18	GRCh38.p7	<- currently shown
NM_005018.2	minus strand	NP_005009.2	reverse	NT_187527.1	GRCh38.p7	View SNP on GeneModel
XM_017004293.1	minus strand	XP_016859782.1	reverse	NT_005403.18	GRCh38.p7	View SNP on GeneModel
XM_006712573.2	minus strand	XP_006712636.1	reverse	NT_005403.18	GRCh38.p7	View SNP on GeneModel

Clinical Source in gene region cSNP has frequency double hit refresh

gene model Contig Label Contig mRNA protein mRNA orientation transcript SNP count
(contig mRNA transcript): GRCh38.p7 NT_005403.18 NM_005018.2 NP_005009.2 reverse minus strand 298, coding

Region	Chr. position	mRNA pos	dbSNP rs# cluster id	Heterozygosity	Validation	MAF	Allele origin	3D	Clinically Associated	Clinical Significance	Function	dbSNP allele	Protein residue	Codon pos	Amino acid pos	PubMed
241851063	930	rs1350210358	N.D.							missense	T	Phe [F]	1	288		
241851063	930	rs1452312939	N.D.							contig reference	C	Leu [L]	1	288		
241851068	925	rs547824816	0.000	0.0002						frame shift	-	Ser [S]	1	288		
241851068	925	rs547824816	0.000	0.0002						contig reference	C	Leu [L]	1	288		
241851068	925	rs547824816	0.000	0.0002						nonsense	A		2	286		
241851068	925	rs547824816	0.000	0.0002						contig reference	G	Trp [W]	2	286		

見やすいとは言い難い -> Variant Viewer

dbSNPの遺伝子ビュー(Variation Viewer)

NCBI Resources How To Sign in to NCBI

Variation Viewer

Homo sapiens: GRCh38.p12 (GCF_000001405.38) Chr 2 (NC_000002.12): 241,848,978 - 241,859,811

Pick Assembly Search

Region PCD1 NM_005018.2 Gene Transcript Exons: click an exon above to zoom in, mouse over to see details

Reset All Share this page FAQ Help Version 2.1.4

YouTube

Search examples: PCD1

Genes Other features

Name Location

PCD1 Chr2: 241,849,881 - 241,858,908
PCD1 NT_187527.1: 61,979 - 71,004
C274 Chr9: 5,450,503 - 5,470,567
PCD1LG2 Chr9: 5,510,438 - 5,571,282
VSR Chr10: 71,747,556 - 71,773,580
TNF Chr6: 31,575,567 - 31,578,336
TNF NT_13891.3: 3,053K - 3,056K
TNF NT_167244.2: 2,909K - 2,911K

User Data and Track Hubs History

Assembly Region Details

Features of Interest Other sequence representations for assembly region(s) in view

Sequence ID	Type
Region REGION120	
NT_187523.1	alt
NT_187527.1	alt
NT_187647.1	alt

2 GRC genome issues in this view. Add Track

Variation Data

Filter by

Source database dbSNP (2893) dbVar (404)

In ClinVar Yes (107) No (3190)

Most severe clinical significance pathogenic (66) pathogenic-likely-pathogenic (0) likely-pathogenic (8) drug-response (0) confers sensitivity (0) More...

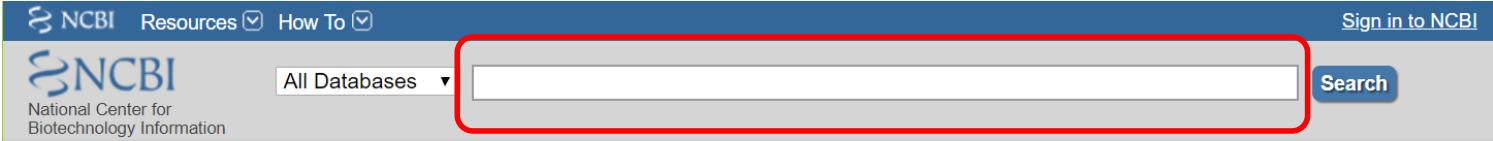
Variant type single nucleotide variant (2700)

Variant ID Location Variant type Gene Molecular consequences Most severe clinical significance 1000G MAF GO-ESP MAF ExAC MAF Publications

nsv429565 12,784 - 242,147,305 copy number variation LOC112268441 and 3863 more Not-Provided 1
nsv3885544 14,238 - 242,106,609 copy number variation LOC112268441 and 3861 more Pathogenic 1
nsv3874648 15,672 - 242,157,305 copy number variation LOC112268441 and 3863 more Pathogenic 1
nsv1004728 36,551 - 242,063,877 copy number variation LOC112268441 and 3860 more Pathogenic 1
nsv535502 36,551 - 242,063,877 copy number variation LOC112268441 and 3860 more Pathogenic 1
nsv2772356 50,659,294 - 242,160,331 copy number variation LOC112268441 and 3094 more Pathogenic 1
nsv3910630 50,659,294 - 242,160,331 copy number variation LOC112268441 and 3094 more Pathogenic 1
nsv2779041 110,076,072 - 242,160,331 copy number variation DPP10 and 2011 more Uncertain-Significance 1
nsv3923963 110,076,072 - 242,160,331 copy number variation DPP10 and 2011 more Uncertain-Significance 1
nsv2770864 130,387,022 - 242,160,331 copy number variation SPAG16 and 1698 more Uncertain-Significance 1
nsv3924199 130,387,022 - 242,160,331 copy number variation SPAG16 and 1698 more Uncertain-Significance 1
nsv1008042 183,229,089 - 242,063,877 copy number variation LOC105376755 and 982 more 1
nsv536073 183,229,089 - 242,063,877 copy number variation LOC105376755 and 982 more 1
nsv3890989 188,818,195 - 242,065,208 copy number variation PARD3B and 933 more Pathogenic 1
nsv531671 188,818,195 - 242,065,208 copy number variation PARD3B and 933 more Pathogenic 1

dbSNPおよびdbVar, ClinVarの情報を genome browser上で確認可能
自分の持つ情報をUploadして併せて表示も可能。

dbSNP以外からの利用方法



The screenshot shows the NCBI homepage. At the top, there is a navigation bar with links for NCBI Resources, How To, and Sign in to NCBI. Below the navigation bar is the NCBI logo and the text "National Center for Biotechnology Information". A search bar is prominently displayed, with a red box highlighting the search input field. To the right of the search bar is a "Search" button. On the left side of the page is a sidebar with a "Resource List (A-Z)" menu containing links to various databases: NCBI Home, All Resources, Chemicals & Bioassays, Data & Software, DNA & RNA, Domains & Structures, Genes & Expression, Genetics & Medicine, Genomes & Maps, Homology, Literature, Proteins, Sequence Analysis, Taxonomy, Training & Tutorials, and Variation. The main content area features a "Welcome to NCBI" section with a brief introduction and links to About the NCBI, Mission, Organization, and NCBI News & Blog. Below this are six sections: Submit (Deposit data or manuscripts into NCBI databases), Download (Transfer NCBI data to your computer), Learn (Find help documents, attend a class or watch a tutorial), Develop (Use NCBI APIs and code libraries to build applications), Analyze (Identify an NCBI tool for your data analysis task), and Research (Explore NCBI research and collaborative projects). To the right of the main content is a "Popular Resources" sidebar listing links to PubMed, Bookshelf, PubMed Central, BLAST, Nucleotide, Genome, SNP, Gene, Protein, and PubChem. At the bottom right, there is a "NCBI News & Blog" section with several news items.

NCBIのWeb site (<https://www.ncbi.nlm.nih.gov/>) から
All Databasesを対象に遺伝子名(例: CYP2D6)で検索すると、、、

NCBIの統合検索機能を利用した情報の取得

NCBI Databases

Results found in 31 databases for: CYP2D6

Literature

Bookshelf	615
MeSH	5
NLM Catalog	11
PubMed	7,058
PubMed Central	8,971

Genes

Gene	84
GEO DataSets	12
GEO Profiles	50,222
HomoloGene	2
PopSet	12
UniGene	10

Genetics

ClinVar	70
dbGaP	0
dbSNP	3,122
dbVar	302
GTR	83
MedGen	48
OMIM	17

Proteins

Conserved Domains	0
Identical Protein Groups	14
Protein	579
Protein Clusters	0
Sparcle	2
Structure	24

Genomes

Assembly	0
BioCollections	0
BioProject	7
BioSample	54
Genome	7
Nucleotide	1,706
Probe	101
SRA	98
Taxonomy	0

Chemicals

BioSystems	210
PubChem BioAssay	3,822
PubChem Compound	1
PubChem Substance	357

[ハンズオン1] dbSNP

[basic]

- ・「rs75961395」と関連が報告された形質は？
- ・「rs75961395」のアリル頻度はどのようなプロジェクトから報告されている？

[advanced]

- ・「rs75961395」はどのプラットフォームで検知可能かを調べましょう

「rs75961395」と関連が報告された形質は？

Google 検索結果
rs75961395

約 249 件 (0.28 秒)

[rs75961395 RefSNP Report - dbSNP - NCBI](https://www.ncbi.nlm.nih.gov/snp/rs75961395)

<https://www.ncbi.nlm.nih.gov/snp/rs75961395> このページを訳す

Sequence name, Change. GRCh37.p13 chr 7, NC_000007.13:g.117149177G>T. GRCh37.p13 chr 7, NC_000007.13:g.117149177G>A. GRCh38.p12 chr 7, NC_000007.14:g.117509123G>T. GRCh38.p12 chr 7 ...

<https://www.ncbi.nlm.nih.gov/snp/rs75961395>

rs75961395

Current Build 152
Released October 2, 2018

Organism	Homo sapiens	Clinical Significance Reported in ClinVar
Position	chr7:117509123 (GRCh38.p12) ?	Gene : Consequence CFTR : Missense Variant
Alleles	G>A / G>T	Publications 8 citations
Variation Type	SNV Single Nucleotide Variation	Genomic View See rs on genome
Frequency	A=0.00004 (11/245624, GnomAD) A=0.00006 (8/125568, TOPMED) A=0.00007 (8/121180, ExAC) (+ 1 more)	

Variant Details

Allele: A (allele ID: 22182)		
ClinVar Accession	Disease Names	Clinical Significance
RCV000007563.8	Cystic fibrosis	Pathogenic
RCV000224170.1	not provided	Pathogenic

Allele: T (allele ID: [68178](#))

ClinVar Accession	Disease Names	Clinical Significance
RCV000577243.1	Cystic fibrosis	Not-Provided

Cystic fibrosis
(囊胞性線維症)

22

<https://biosciencedbc.jp/>

CC BY

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「rs75961395」のアリル頻度はどのようなプロジェクトから報告されている？

Frequency A=0.00004 (11/245624, GnomAD)
 A=0.00006 (8/125568, TOPMED)
 A=0.00007 (8/121180, ExAC) (+ 1 more)

Variant Details Filter:

Study	Population	Group	Sample Size	Ref Allele	Alt Allele
gnomAD - Exomes	Global	Study-wide	245624	G=0.99996	A=0.00004
gnomAD - Exomes	European	Sub	133452	G=0.99992	A=0.00008
gnomAD - Exomes	Asian	Sub	48022	G=1.0000	A=0.0000
gnomAD - Exomes	American	Sub	33550	G=1.0000	A=0.0000
gnomAD - Exomes	African	Sub	15286	G=1.0000	A=0.0000
gnomAD - Exomes	Ashkenazi Jewish	Sub	9842	G=1.00	A=0.00
gnomAD - Exomes	Other	Sub	5472	G=1.00	A=0.00
TopMed	Global	Study-wide	125568	G=0.99994	A=0.00006
ExAC	Global	Study-wide	121180	G=0.99993	A=0.00007
ExAC	Europe	Sub	73272	G=0.9999	A=0.0001
ExAC	Asian	Sub	25146	G=1.0000	A=0.0000
ExAC	American	Sub	11534	G=1.0000	A=0.0000
ExAC	African	Sub	10320	G=1.0000	A=0.0000
ExAC	Other	Sub	908	G=1.00	A=0.00
gnomAD - Genomes	Global	Study-wide	30954	G=1.0000	A=0.0000
gnomAD - Genomes	European	Sub	18480	G=0.9999	A=0.0001
gnomAD - Genomes	African	Sub	8734	G=1.000	A=0.000
gnomAD - Genomes	East Asian	Sub	1618	G=1.000	A=0.000
gnomAD - Genomes	Other	Sub	982	G=1.00	A=0.00
gnomAD - Genomes	American	Sub	838	G=1.00	A=0.00
gnomAD - Genomes	Ashkenazi Jewish	Sub	302	G=1.00	A=0.00

「rs75961395」はどのプラットフォームで検知可能かを調べましょう

Variant Details	48 SubSNP, 4 Frequency, 3 ClinVar submissions	
Clinical Significance	Filter: <input type="text"/>	
Frequency		
Aliases		
Submissions		
History		
Publications		
	No	Submitter
	4	TopMed
	5	gnomAD - Exomes
	6	gnomAD - Genomes
	7	ExAC
	8	ILLUMINA ss3654179603
	9	ILLUMINA ss3653307594
	10	ILLUMINA ss3644951976
	11	ILLUMINA ss3640845825

Custom AFFY chips

dbSNP Short Genetic Variations

dbVar ClinVar GaP PubMed Nucleotide Protein

Search small variations in dbSNP or large structural variations in dbVar

Search Entrez for Go

Method Detail

Submitter Method Handle: ILLUMINA
 Submitter Method ID: AFFY-CHIP
 Submitted method description: This method is created specially for custom AFFY chips

This method was used in the following submission:

Submitter Handle	Batch Type	Submitter batch id	Release build id
ILLUMINA	Assay	Axiom_BioBank1	151

多型データベースの紹介 2: ExAC/gnomAD

ExAC (Exome Aggregation Consortium)

- 運営元：米国ブロード研究所
- URL: <http://exac.broadinstitute.org/>
- 様々のプロジェクトからの60,706名の全エクソームシークエンス(WES)データを併せて解析
 - 日本人は76名分のみ
- 次世代シークエンサーにおけるCoverageの情報トラックあり
- 7つのpopulationにおけるアリル頻度の違いを表示

ExACの画面説明、操作

ExAC Browser Beta About Downloads Terms Contact Jobs FAQ

Interested in working on the development of this resource? [Apply here.](#)

ExAC Browser (Beta) | Exome Aggregation Consortium

Search for a gene or variant or region

Examples - Gene: [PCSK9](#), Transcript: [ENST00000407236](#), Variant: [22-46615880-T-C](#), Multi-allelic variant: [rs1800234](#), Region: [22:46615715-46615880](#)

About ExAC

The [Exome Aggregation Consortium](#) (ExAC) is a coalition of investigators seeking to aggregate and harmonize exome sequencing data from a wide variety of large-scale sequencing projects, and to make summary data available for the wider scientific community.

The data set provided on this website spans 60,706 unrelated individuals sequenced as part of various disease-specific and population genetic studies. The ExAC Principal Investigators and groups that have contributed data to the current release are listed [here](#).

All data here are released under a [Fort Lauderdale Agreement](#) for the benefit of the wider biomedical community - see the terms of use [here](#).

Sign up for our mailing list for future release announcements [here](#).

Recent News

August 8, 2016
- CNV calls are now available on the ExAC browser

March 14, 2016
- Version 0.3.1 ExAC data and browser (beta) is released! ([Release notes](#))

January 13, 2015
- Version 0.3 ExAC data and browser (beta) is released! ([Release notes](#))

October 29, 2014

gnomAD (The Genome Aggregation Database、ノマド)

- 運営元: ブロード研究所
- URL: <https://gnomad.broadinstitute.org/>
- ExACの後継プロジェクト
 - 125,748名の全エクソームシークエンスデータ
 - 15,708名の全ゲノムシークエンスデータ
- ExACよりも、より細かい条件での頻度情報を取得できる
- 同じ解析手法により解析している為、dbSNPより高精度

gnomADの画面説明、操作



gnomAD browser

Search by gene, region, or variant

About Downloads Terms Contact Jobs FAQ

This is a new version of the gnomAD browser. The old version is available at <http://gnomad-old.broadinstitute.org>

gnomAD

genome aggregation database

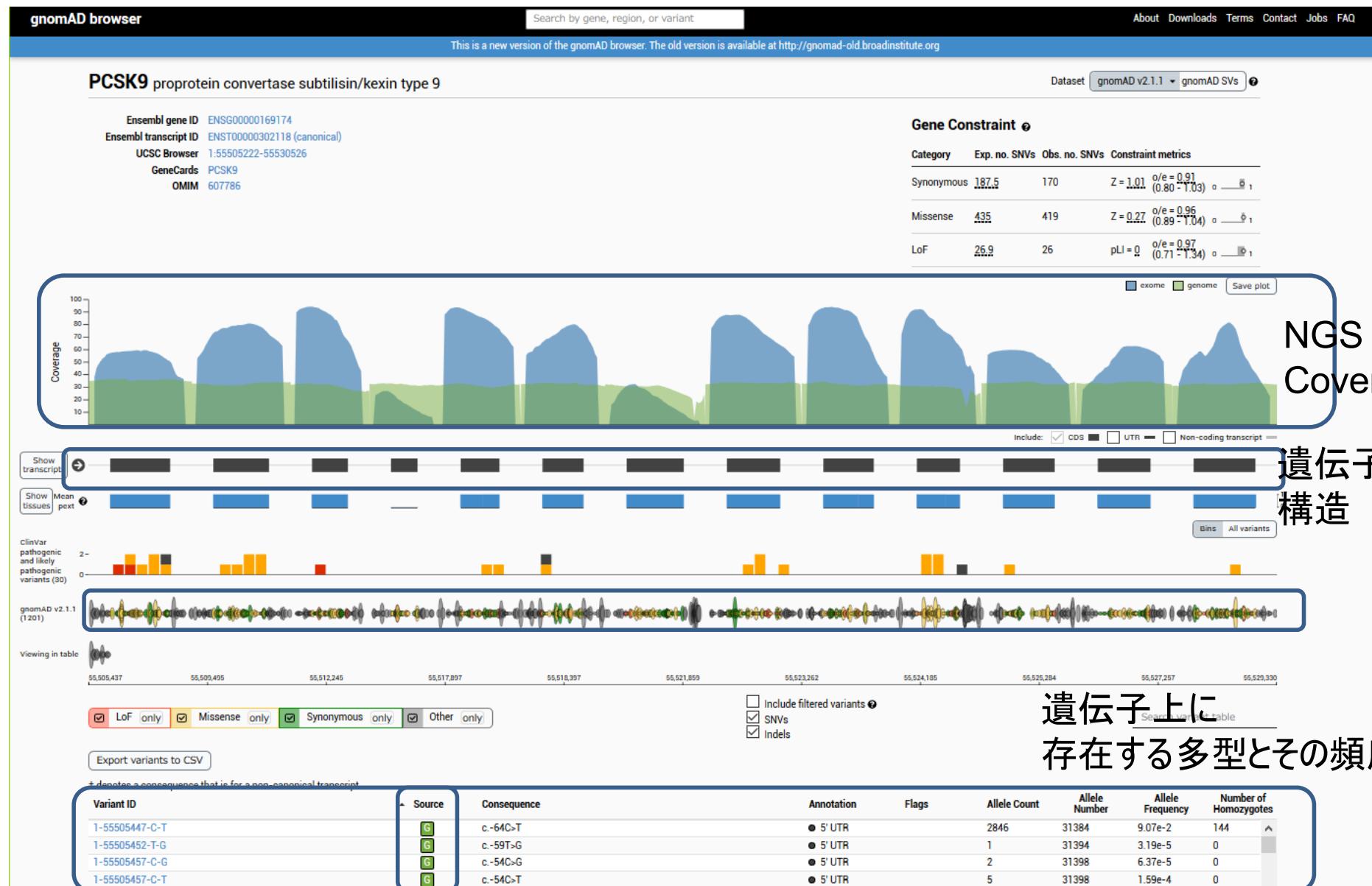
Search by gene, region, or variant

Examples - Gene: PCSK9, Variant: 1-55516888-G-GA

The genome Aggregation Database (gnomAD) is a resource developed by an international coalition of researchers who have aggregated, summarizing, genome sequencing data from a wide variety of large-scale sequencing projects, and making summary data available for the wider scientific community.

The data set provided on this website spans 125,748 exome sequences and 15,708 whole-genome sequences from unrelated individuals sequenced as part of various disease specific and population

gnomADの検索結果(遺伝子)



gnomADの検索結果(バリアント)

This is a new version of the gnomAD browser. The old version is available at <http://gnomad-old.broadinstitute.org>

Insertion: 1-55505552-A-ACTG

Filter	Exomes	Genomes	Total
Allele Count	Pass	Pass	24095
Allele Number	19607	4488	192344
Allele Frequency	0.1217	0.1436	0.1253
Popmax Filtering AF (95% confidence)	0.1723	0.1994	

This variant is multiallelic. Other alt alleles are:

- 1-55505552-A-AATGCTG
- 1-55505552-A-ACTGCTG
- 1-55505552-A-ACTGCTGCTG
- 1-55505552-A-ACTGCTGCTGCTG
- 1-55505552-ACTG-A
- 1-55505552-ACTGCTG-A

Annotations

This variant falls on 2 transcripts in 1 gene.

inframe insertion

- PCSK9
 - ENST00000302118 *
HGVS: p.Leu23dup
 - ENST00000452118
HGVS: p.Leu23dup

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
African	3359	17352	306	0.1936
Ashkenazi Jewish	1417	8730	82	0.1623
South Asian	3250	23124	183	0.1405
European (non-Finnish)	10076	78928	489	0.1277
Other	665	5572	21	0.1193
Overall	1556	13538	61	0.1149
Korean	297	2562	13	0.1159
East Asian	1073	9380	43	0.1144
Japanese	2	40	0	0.05000
Male	835	7092	33	0.1177
Female	721	6446	28	0.1119
European (Finnish)	1736	18904	57	0.09183
Latino	2036	26196	52	0.07772
Total	24095	192344	1251	0.1253

Include: Exomes Genomes

Dataset gnomAD v2.1.1

- gnomAD v2.1.1 (141,456 samples)
- gnomAD v2.1.1 (controls) (60,146 samples)
- gnomAD v2.1.1 (non-cancer) (134,107 samples)
- gnomAD v2.1.1 (non-neuro) (114,704 samples)
- gnomAD v2.1.1 (non-TOPMed) (136,743 samples)

Report

- Report this variant
- Request additional information

Age Distribution

Heterozygotes Homozygotes

Population毎、性別毎の頻度情報
・ExACよりも粒度が細かい

頻度の再計算が可能

- ・全サンプル
- ・コントロールサンプル
- ・非がんサンプル
- ・非神経疾患サンプル
- ・TOPMedに含まれないサンプル

Trans-Omics for Precision Medicine Program (TOPMed)

[ハンズオン2] gnomAD

[basic]

- 2番染色体の位置242794191はどのようなアリルが報告されていますか？
- 上記の多型が存在する遺伝子の名前を調べましょう。

[advanced]

- rs1801133は、東アジア集団において男女のアリル頻度に差があるか、また、非がんサンプルと非神経疾患サンプルでアリル頻度に差があるか調べましょう。

2番染色体の位置242794191はどのようなアリルが報告されていますか？

https://gnomad.broadinstitute.org/region/2-242794191

gnomAD v2.1.1
(11)

Viewing in table

242,794,171 242,794,175 242,794,179 242,794,184 242,794,188 242,794,193 242,794,197 242,794,202 242,794,206 242,794,211

LoF only Missense only Synonymous only Other only

Exomes SNVs
 Genomes Indels Filtered variants ?

Search variant table

Export variants to CSV

Variant ID	Source	Consequence	Annotation	Flags	Allele Count	Allele Number
2-242794173-T-A	E G	c.593-38A>T	● intron		201	274816
2-242794173-T-C	E	c.593-38A>G	● intron		1	243784
2-242794176-G-A	E	代替アレル(alternative allele)がTの場合とCの場合の2つが報告されているmultiallelic variant				
2-242794177-C-G	E					
2-242794178-C-T	E					
2-242794179-G-A	E G	c.593-44C>T	● intron		91	268924
2-242794189-G-GT	G	c.593-55dupA	● intron		2	30606
2-242794190-T-TG	G	c.593-56dupC	● intron		1227	28396
2-242794191-G-T	G	c.593-56C>A	● intron		1	28632
2-242794191-G-C	G	c.593-56C>G	● intron		1240	28632
2-242794193-G-C	G	c.593-58C>G	● intron		3	31034

上記の多型が存在する遺伝子の名前を調べましょう。

gnomAD browser Search by gene, region, or variant About Downloads Terms Contact
This is a new version of the gnomAD browser. The old version is available at <http://gnomad-old.broadinstitute.org>

Single nucleotide variant: 2-242794191-G-C

Dataset gnomAD v2.1.1 ▾ ?

Filter	Exomes	Genomes	Total	References
No variant	Pass			<ul style="list-style-type: none"> dbSNP (rs55829775) UCSC
Allele Count	1240	1240		
Allele Number	28632	28632		
Allele Frequency	0.04331	0.04331		
Popmax Filtering AF ⓘ (95% confidence)	0.05418			

This variant is multiallelic. Other alt alleles are:

- [2-242794191-G-T](#)

Annotations

This variant falls on 3 transcripts in 1 gene.

intron

- PDCD1
- [ENST00000334409 *](#)
- [ENST00000343705](#)
- [ENST00000418831](#)

PDCD1 (別名 : PD-1)

Population Frequencies

Allele Allele Number of www.ncbi.nlm.nih.gov/projects/SNP/snp_ref.cgi?rs=rs55829775

Allele Frequency

Age Distribution

rs1801133は、東アジア集団において男女のアリル頻度に差があるか。

← → C https://gnomad.broadinstitute.org/variant/1-11856378-G-A

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
Latino	17788	35440	4654	0.5019
Ashkenazi Jewish	4740	10370	1089	0.4571
European (non-Finnish)	43635	129108	7511	0.3380
Other	2325	7220	395	0.3220
Overall	5790	19944	929	0.2903
Korean	1671	3818	366	0.4377
Japanese	49	152	8	0.3224
East Asian	3623	14424	486	0.2512
Male	2946	10086	465	0.2921
Female	2844	9858	464	0.2885
European (Finnish)	3799	23116	694	0.2909
South Asian	4443	30616	389	0.1451
African	2714	24970	158	0.1087
Female	40861	129446	7576	0.3157
Male	46373	153338	8243	0.3024
Total	87234	282784	15819	0.3085

Include: Exomes Genomes

Genotype Quality Metrics

→差はない

<https://gnomad.broadinstitute.org/variant/1-11856378-G-A>

非がんサンプルと非神経疾患サンプルでアリル頻度に差があるか調べましょう → 差がない

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
Latino	17647	35108	4626	0.5026
Ashkenazi Jewish	4500	9862	1031	0.4563
European (non-Finnish)	39926	118098	6877	0.3381
Other	2165	6700	372	0.3231
East Asian	5620	19242	906	0.2921
European (Finnish)	5794	25100	693	0.2308
South Asian	4427	30526	388	0.1450
African	2562	23616	151	0.1085
Female	38392	121576	7176	0.3158
Male	44249	146676	7868	0.3017
Total	82641	268252	15044	0.3081

非がん

https://gnomad.broadinstitute.org/variant/1-11856378-G-A?dataset=gnomad_r2_1_non_cancer

Total: 0.3081

East Asian: 0.2921

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
Latino	15735	31078	4180	0.5063
Ashkenazi Jewish	2980	6458	693	0.4614
European (non-Finnish)	35207	103102	6124	0.3415
Other	1792	5592	312	0.3205
East Asian	4455	14966	747	0.2977
European (Finnish)	4155	17894	519	0.2322
South Asian	4442	30608	389	0.1451
African	2102	19602	117	0.1072
Female	33421	105204	6322	0.3177
Male	37447	124096	6759	0.3018
Total	70868	229300	13081	0.3091

非神経疾患

https://gnomad.broadinstitute.org/variant/1-11856378-G-A?dataset=gnomad_r2_1_non_neuro

Total: 0.3091

East Asian: 0.2977

多型データベースの紹介 3: jMorp、HGVD

Japanese Multi Omics Reference Panel (jMorp) とは

- 運営元： 東北メディカル・メガバンク機構
- URL:
<https://jmorp.megabank.tohoku.ac.jp/201905/>
- 日本人の約3,500名の住民コホートからの
全ゲノムシーケンスデータから
- 以前は、iJGVDにて公開。

jMorpの画面説明、操作

The screenshot shows the homepage of the Japanese Multi Omics Reference Panel (jMorp). At the top, there is a navigation bar with links for Sequence, Variation, Proteome, Metabolome, Repository, Downloads, Help, and a link to the Licht Toyo-oka project. Below the navigation bar, there is a green banner with two hands and the text "Welcome to Japanese Multi Omics Reference Panel". On the left side, there is a sidebar with icons and labels for Phenome, Metabolome, Proteome, Transcriptome, Methylome, Genome Variation, and Genome Sequence. The "Genome Variation" section is highlighted with a blue border. On the right side, there is a box for "jMorp release 201905" dated May 8th, 2019, which released a 3.5KJPv2 Genotype Frequency dataset from 3.5K Japanese individuals. There is also a section for "jMorp Publication" listing research papers by Tadaka et al. at Iwate Medical Megabank Organization.

jMorp

Sequence | Variation | Proteome | Metabolome | Repository | Downloads | Help | Licht Toyo-oka

Welcome to
Japanese Multi Omics
Reference Panel.

Phenome
To be provided

Metabolome

Proteome

Transcriptome
Iwate Medical Megabank Organization; iMethyl

Methylome
Iwate Medical Megabank Organization; iMethyl

Genome Variation

Genome Sequence

jMorp release 201905

May 8th, 2019
3.5KJPv2 Genotype Frequency dataset
We released 3.5KJPv2 Genotype Frequency dataset calculated from 3.5K Japanese individuals. You can download it from [Downloads](#).

[More](#)

jMorp Publication

Tadaka S, Saigusa D, Motoike IN, Inoue J, Aoki Y, Shirota M, Koshiba S, Yamamoto M, Kinoshita K.
"jMorp: Japanese Multi Omics Reference Panel"
Nucleic Acids Research. 2018 Jan 4;46(D1):D551-D557. [\[PubMed\]](#)

Tadaka S, Katsuoka F, et al.,
"3.5KJPv2, An allele frequency panel of 3,552 Japanese Individuals"
bioRxiv 529529; doi: <https://doi.org/10.1101/529529> [\[bioRxiv\]](#)

[More](#)

jMorp release 201905 / LastUpdate: May 8th, 2019 / Conditions of Use
Tohoku Medical Megabank Organization, Tohoku University [\[link\]](#)

jMorpの検索方法

The screenshot shows the jMorp search interface. At the top, there is a navigation bar with links for Sequence, Variation, Proteome, Metabolome, Repository, Downloads, Help, and a user account. Below the navigation bar, the jMorp logo and a subtitle 'jMorp ~Japanese Multi Omics Reference Panel~' are displayed. The main search area is titled 'Genomic Variants'. It features three search input fields: 'Search by gene name', 'Search by rs#', and 'Search by region (GRCh37/hg19)'. A large blue arrow points upwards from the search results area towards the search input fields. Below the search fields, there is a text input field labeled 'Gene name' with examples: ALDH2, NFE2L2, GATA1. To the right of the search input fields is a 'Search' button. At the bottom of the page, there is a footer with copyright information: 'jMorp release 201806 / LastUpdate: May 8th, 2018 / Conditions of Use' and 'Tohoku Medical Megabank Organization, Tohoku University'.

遺伝子名、rs番号、領域を指定し、検索可能

jMorpの検索結果

jMorp -Japanese Multi Omics Reference Panel-

Search by gene: ALDH2 GRCh37/hg19

Showing unfiltered version.

4148 variants found

Filter by keyword

ToMMoにおける頻度

gnomADにおける頻度

Type	Position	Ref/Alt	rs#	Annotation	MeanDepth (162PE)	Gene	ToMMo 3.5KJPNv2 Allele Frequency Panel	gnomAD AFR.	gnomAD AMR.	gnomAD ASJ.	gnomAD EAS.	gnomAD NFE.
SNV	12:112199811	A/T		upstream_gene_variant	22.0/22.0	ALDH2	0.0001					
SNV	12:112199828	G/T	rs1022625657	upstream_gene_variant	21.9/21.9	ALDH2	0.0010					
SNV	12:112199876	C/T	rs944612066	upstream_gene_variant	22.5/22.5	ALDH2	0.0001					0.0001
SNV	12:112199884	T/C	rs539004647	upstream_gene_variant	22.4/22.4	ALDH2	0.0006					
SNV	12:112199912	G/A	rs914760172	upstream_gene_variant	22.3/22.3	ALDH2	0.0006					

jMorpの検索結果 (タンパク質立体構造上へのマッピング)

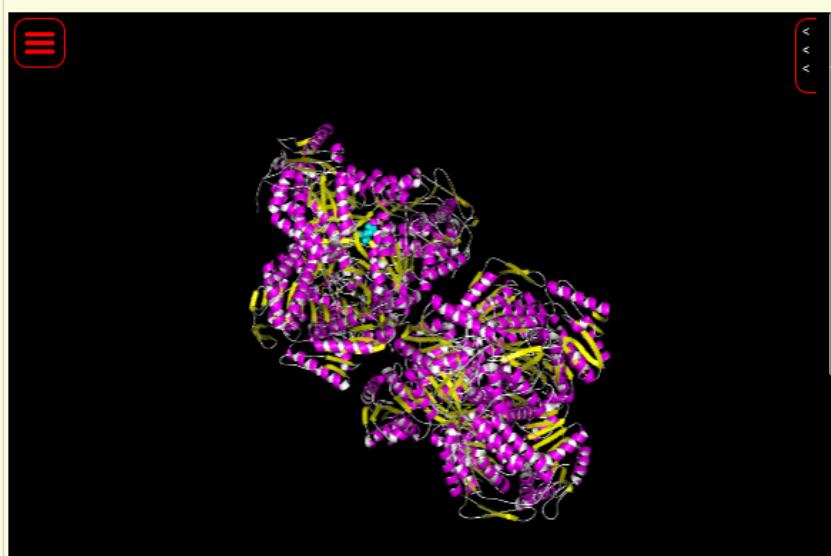
Type	Position	Ref/Alt	rs#	Annotation	Gene	MeanDepth (162PE)	JPA	ToMMo 3.5KJPNv2 Allele Frequency Panel	gnomAD AFR.	gnomAD AMR.	gnomAD ASJ.	gnomAD EA\$.	gnomAD NFE.	
SNV	12:112241766	G/A	rs671	missense_variant (p.Glu504Lys)	ALDH2	18.4/18.4	V1&V2		0.1959	0.0002	0.0012		0.2671	

BlastHit1 BlastHit2 BlastHit3 BlastHit4 BlastHit5 BlastHit6 BlastHit7 BlastHit8 BlastHit9 BlastHit10

Blast result

mRNA info: Homo sapiens aldehyde dehydrogenase 2 family (mitochondrial) (ALDH2), transcript variant 1, mRNA.
mRNA change: NM_000690.3:c.1510G>A_NP_000681.2:p.504E>K
Query: ALDH2 (GeneID: 217) | NM_000690.3 | NP_000681.2
Subject: 5I13
e-value: 0.0000
Sequence Identity: 100.00

5I13_entity_1 (1/1)



Chain:A (1/8)

Single

- Secondary Structure: E
- Relative ASA: 0.374

BioUnit:1

- delta Relative ASA: 0.295

BioUnit:2

- delta Relative ASA: NA

Mapped Position

- auth_asym_id: A, auth_seq_id: 487
- label_asym_id: A, label_seq_id: 504

Human Genome Variation Database (HGVD)とは

- 運営元： 京都大学
- URL:
<http://www.hgvd.genome.med.kyoto-u.ac.jp/>
- 1208名の全エクソームシークエンスデータからの多型情報 および 3,248名のSNP chipからの多型情報
- 長浜コホートからのサンプルが中心

HGVDの画面説明、操作

Human Genetic Variation Database

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Total downloads: 6,259

Welcome to Human Genetic Variation Database

Search database

Gene name/ID GO

dbSNP rsID GO

Pathogenic Variation GO

Chromosome
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y

What's New?

- ▶ 08/02/2017 HGVD version 2.3 is now downloadable.
- ▶ 04/17/2017 New HLA typing software 'HLA-HD' is released. [link](#)