

遺伝子のバリエーション

清水謙多郎

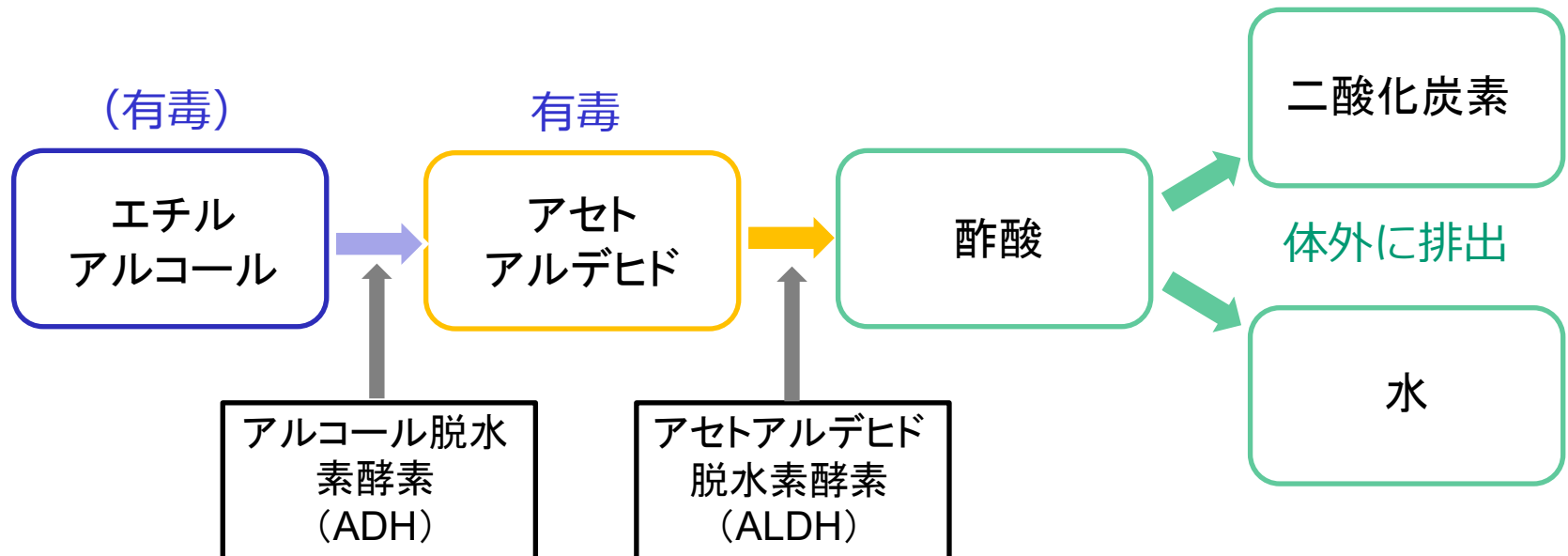
shimizuk@fc.jwu.ac.jp

SNPとバリエーション

- **SNP**（スニップ）とは、一塩基多型（Single Nucleotide Polymorphism）の略
 - 同じ生物種内で、ゲノム配列中の1塩基が個体によって異なる現象
 - その変異が一定（一般に1%）以上の頻度で集団中に存在するもの
- ヒトではおよそ300万か所以上のSNPが知られている
- タンパク質の構造や機能に影響を及ぼす可能性があり、例えば、体質・病気の進行・薬の効き方の違いなどを説明できることがある
- **バリエーション**（variant）：DNA配列に見られる塩基の違い（変異・変化）を総称した用語
 - SNPのような1塩基の違いだけでなく、小さな挿入・欠失（indel）やコピー数多型（CNV）も含む広い概念
 - 以前は頻度1%以上のものを多型（SNP）として区別していたが、その境界は明確ではなく、現在では境界はあいまいで、臨床遺伝学やゲノム解析ではバリエーションが包括的に使われる

飲酒に関係する重要な酵素とその遺伝子

- **ALDH**: アセトアルデヒド脱水素酵素（およびその遺伝子）
 - エチルアルコールの代謝によって生じるアセトアルデヒドを酢酸に分解する酵素
 - **ALDH2**は、アセトアルデヒドの解毒の大部分を担う
- **ADH**: アルコール脱水素酵素（およびその遺伝子）
 - エチルアルコールを分解してアセトアルデヒドにする酵素
 - レチノール、ステロイド、脂肪酸などを分解する型の酵素もある
 - **ADH1B**は、エチルアルコールの分解に重要





生活習慣病予防

健康寿命

身体活動・運動

栄養・食生活

休養・こころの健康

歯・口腔の健康

飲酒

喫煙

感覚器など

健康政策

健康用語辞典

執筆者一覧

e-ヘルスネット > 飲酒 > アルコールと依存 > アルコール依存症と遺伝



アルコール依存症と遺伝

アルコール依存症の原因に遺伝が関係することは確かです。特にアルコールを分解する酵素の遺伝子による違いが、依存症の発症に関与しています。

2. アルコール代謝酵素遺伝子と依存症の関係

数多くの遺伝子が原因の候補として検討されましたが、アルコールを代謝する酵素の遺伝子以外に決定的な候補は見つかっていません。

アルコールを代謝する酵素の遺伝子にはいくつかのタイプ（遺伝子多型）があり、依存症に関係します。肝臓では、アルコールをアセトアルデヒドに分解するアルコール脱水素酵素(ADH1B)と、アルコールが代謝されてきた有害なアセトアルデヒドを無毒な酢酸に分解するアルデヒド脱水素酵素(ALDH2)がアルコール代謝の中心的な役割を果たしますが、その両方の遺伝子に多型が存在します。

ADH1BにはHis48Arg多型があり、48番目のアミノ酸がヒスチジン(His)の人とアルギニン(Arg)の人がいます。Hisの人は東洋人に多く、アルコールを分解する速度が非常に速いという特徴があります。

またALDH2遺伝子にはGlu487Lys多型があり、ALDH2の487番目のアミノ酸がリシン(Lys)の人とグルタミン酸(Glu)の人がいます。Lysの場合にはALDH2酵素は働かなくなります。

ADH1B遺伝子がHisの人は酵素がよく働くために飲酒するとアセトアルデヒドが早くでき、ALDH2遺伝子がLysの人は飲酒してできたアセトアルデヒドがなかなか分解されずに体内に貯留するので、飲酒すると顔が赤くなったり動悸がしたりして不快な反応を引き起こして依存症にはなりづらくなります。

3. 遺伝子と環境因子の相互作用

依存症の原因に環境が関係することは「アルコール依存症の危険因子」で述べましたが、同じ環境におかれても依存症になる人とならない人がいます。その原因に遺伝子が関係しているという説があります。これを遺伝子と環境の相互作用と言います。

例えば、上述のALDH2酵素が働かないタイプの遺伝子をもった人は依存症になりにくいことが分かっていますが、アルコール依存症の人の中で、この遺伝子を持っている人の割合を調べると、1979年には2.5%でしたが、この割合は時代とともに変化して、最近（2006年から2010年）では15.4%と高くなっています^[6]。これは、環境の変化によって、本来は依存症になりにくい遺伝子を持った人も、依存症になってしまうことを示唆していると考えられます。

関連キーワード

1B型アルコール脱水素酵素 >

2型アルデヒド脱水素酵素 >

アルコール依存症 >


アルコール依存症の危険因子 >


アルコール依存症の心理・社会的治療

女性アルコール依存症者の摂食障害

NCBIのサイトにアクセス

- アメリカ国立生物工学情報センター
(National Center of Biotechnology Information, NCBI)
 - バイオテクノロジーや分子生物学に関連するデータベースやソフトウェアを開発し、サービスを提供している
- <https://www.ncbi.nlm.nih.gov/>

 An official website of the United States government [Here's how you know](#) ▼

 **National Library of Medicine**
National Center for Biotechnology Information Log in

All Databases ▼ Search

NCBI Home

- Resource List (A-Z)
- All Resources
- Chemicals & Bioassays
- Data & Software
- DNA & RNA
- Domains & Structures
- Genes & Expression
- Genetics & Medicine
- Genomes & Maps
- Homology
- Literature
- Proteins


Welcome to NCBI

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

[About the NCBI](#) | [Mission](#) | [Organization](#) | [NCBI News & Blog](#)


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



Popular Resources


- [PubMed](#)
- [Bookshelf](#)
- [PubMed Central](#)
- [BLAST](#)
- [Nucleotide](#)
- [Genome](#)
- [SNP](#)
- [Gene](#)
- [Protein](#)
- [PubChem](#)

[NCBI News & Blog](#)

NCBIのウェブページ

 An official website of the United States government [Here's how you know](#) ✓



 shimizu.jwu@gmail...

All Databases ▾

Search

NCBI Home

Resource List (A-Z)

All Resources

Chemicals & Bioassays

Data & Software

DNA & RNA

Domains & Structures

Genes & Expression

Genetics & Medicine

Genomes & Maps

Homology

Literature

Proteins

Sequence Analysis

Taxonomy

Training & Tutorials

Variation

Welcome to NCBI

The National Center for Biotechnology Information advances science and health by providing access to biomedical and genomic information.

[About the NCBI](#) | [Mission](#) | [Organization](#) | [NCBI News & Blog](#)

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



Research

Explore NCBI research and
collaborative projects



Popular Resources

[PubMed](#)

[Bookshelf](#)

[PubMed Central](#)

[BLAST](#)

[Nucleotide](#)

[Genome](#)

[SNP](#)

[Gene](#)

[Protein](#)

[PubChem](#)

NCBI News & Blog

NCBI Hidden Markov Models (HMM)
Release 16.0 Now Available!

22 Aug 2024

Download release 16.0 of the NCBI
protein profile Hidden Markov models

Quick & Easy Access to Mpx Data
Through NCBI Virus

21 Aug 2024

The World Health Organization
(WHO) declared the recent outbreak of

NCBI's First-Ever BioEd Summit Was a
Success!

19 Aug 2024

「SNP」をクリックする

SNPの検索

An official website of the United States government [Here's how you know](#)

NIH National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP

Advanced 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](#)

i ALFA Project Release 3 with over 900M variants from >200K subjects is now [available](#) (August 3, 2023)

The goal is to provide allele frequency from more than 1 million dbGaP subjects with regular updates. Visit the project [page](#) for more information or view the introduction video below.

ALFA CoLab Presentation, ASHG2020

後で見る 共有

NCBI ALFA

「ALDH2」と入力して
「Search」ボタンをクリック

SNPの検索

手順としては次の次の
スライドに飛びます

An official website of the United States government [Here's how you know](#)

NIH National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP

SNP ALDH2 Search

Create alert Advanced Help

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

Send to: Filters: Manage Filters

Find related data

Database: Select Find items

Search details

ALDH2[All Fields]

Search See more...

Recent activity

Turn Off Clear

ALDH2 (18788) SNP

CYP2D6 cytochrome P450 family 2 subfamily D member 6 [Sus scrofa] Gene

CYP2D6 cytochrome P450 family 2 subfamily D member 6 [Gallus gallus] Gene

CYP2D6 cytochrome P450 family 2 subfamily D member 6 [Homo sapien] Gene

CYP2D6 AND (alive[prop]) (125) Gene

See more...

Search results

Items: 1 to 20 of 18788

<< First < Prev Page 1 of 940 Next > Last >>

☒ rs671 [Homo sapiens]

1.

Variant type: SNV

Alleles: G>A [Show Flanks]

Chromosome: 12:111803962 (GRCh38)
12:112241766 (GRCh37)

Canonical SPDI: NC_000012.12:111803961:G:A

Gene: ALDH2 (Varview)

Functional Consequence: missense_variant, coding_sequence_variant

Clinical significance: risk-factor, pathogenic, drug-response, protective

Validated: by frequency, by alfa, by cluster

MAF: A=0.006055/1921 (ALFA)
A=0./0 (PRJEB36033)
A=0./0 (TWINUK)

...more

HGVS: NC_000012.12:g.111803962G>A, NC_000012.11:g.112241766G>A,
NG_012250.2:g.42076G>A, NM_000690.4:c.1510G>A, NM_000690.3:c.1510G>A,
NM_001204889.2:c.1369G>A, NM_001204889.1:c.1369G>A, NP_000681.2:p.Glu504Lys,
NP_000681.1:p.Glu504Lys

PubMed LitVar

☐ rs13306164 [Homo sapiens]

2.

Variant type: SNV

Alleles: C>T [Show Flanks]

Chromosome: 12:111783251 (GRCh38)
12:112221055 (GRCh37)

Canonical SPDI: NC_000012.12:111783250:C:T

Gene: ALDH2 (Varview)

Functional Consequence: coding_sequence_variant, intron_variant, synonymous_variant

Clinical Significance

benign

drug response

likely benign

pathogenic

protective

risk factor

Validation Status

by-ALFA

by-cluster

by-frequency

Publication

PubMed Cited

PubMed Linked

Function Class

inframe deletion

inframe insertion

initiator codon variant

intron

missense

non coding transcript variant

synonymous

Variation Class

del

delins

ins

mnv

Annotation

somatic

Global MAF

Custom range...

Clear all

多数のSNPの中から、先
頭の「rs671」を選択

ゲノムブラウザ上での表示

Have you explored our new Comparative Genome Viewer (CGV) yet?

Genome Data Viewer

Home Share this page Reset All More Tools More Info

Homo sapiens
(human)

Assembly: GRCh38.p14 (GCF_000001405.40) Chr 12 (NC_000012.12)

Search assembly

ALDH2

Examples

Assemblies

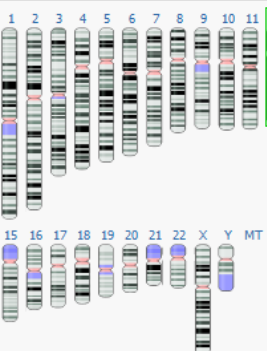
Ideogram View

Unplaced/unlocalized scaffolds:

166

Alt loci/patches:

514



Tracks and User Data

BLAST

Tracks by Accession

Assembly Region Details

History

NC_000012.12: 111,761,873 - 111,822,592

p13.32 p13.31 p13.31 p13.2 p12.3 p12.2 p12.1 p11.22 p11.1 q12 q13.11 q13.12 q13.13 q13.2 q13.3 q14.1 q14.2 q15 q21.1 q21.2 q21.31 q21.32 q21.33 q22 q23.1 q23.2 q23.3 q24.11 q24.12 q24.21 q24.23 q24.31 q24.32 q24.33

Region: ALDH2 Transcript: NM_000690.4 Exons: click an exon to zoom in, mouse over to see details

NC_000012.12: 111,765 K 111,770 K 111,775 K 111,780 K 111,785 K 111,790 K 111,795 K 111,800 K 111,805 K 111,810 K 111,815 K 111,820 K

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

ALDH2 (+4) MIR6761 (+31)

Genes, Ensembl release 110

ENS000000111275 (+) ENS000000257767 ENS000000450353.4 ENS000000450353.4

Cited Variations, dbSNP b156 v2

rs886205 | R/C/G/T rs4767944 | C/G/T rs2238152 | G/T rs7312055 | G/R rs7311852 | C/R/A/G rs10849970 | G/R/C rs441 | T/C rs4646776 | G/A/C rs4646777 | G/R/C rs671 | G/R rs1941669 | T/G rs2158029 | G/A rs7296651 | C/R/G rs16941667 | C/T rs2339840 | C/T

Live RefSNPs, dbSNP b156 v2

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation Rel...

28 482 21084 553 9 25 34295 44639 44349 22130

111,765 K 111,770 K 111,775 K 111,780 K 111,785 K 111,790 K 111,795 K 111,800 K 111,805 K 111,810 K 111,815 K 111,820 K

NC_000012.12: 112M..112M (60,720 nt)

Tracks shown: 8/947

rs671を選択

rs671
Variation ID: rs671
Variation Type: SNV, length 1
Alleles: G/A
[Genomic locations]
GCF_000001405.40: NC_000012.12 @ 111803962
GCF_000001405.25: NC_000012.11 @ 112241766
[Links & Tools]
SNP summary: rs671
ClinVar (2): rs671
PubMed (293): rs671

SNP summaryの表示
のところでrs671を選択

SNPを調べてみよう

dbSNP Short Genetic Variations

Search for terms

Search

Examples: rs268, BRCA1 and more

Advanced search



Welcome to the Reference SNP (rs) Report

All alleles are reported in the [Forward orientation](#). Click on the [Variant Details](#) tab for details on Genomic Placement, Gene, and Amino Acid changes. HGVS names are in the [HGVS](#) tab.

Reference SNP (rs) Report

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

rs671

Current Build 156

Released September 21, 2022

Organism *Homo sapiens*

Position chr12:111803962 (GRCh38.p14) [?](#)

Alleles G>A

Variation Type SNV Single Nucleotide Variation

Frequency
A=0.006055 (1921/317236, ALFA)
A=0.008958 (2371/264690, TOPMED)
A=0.018882 (4582/242666, GnomAD_exome) (+ 17 more)

Clinical Significance Reported in [ClinVar](#)

Gene : Consequence ALDH2 : Missense Variant

Publications 293 citations

[LitVar²](#) 804

Genomic View [See rs on genome](#)

「Variant Details」

Frequency

Variant
Details

Clinical
Significance

HGVS

Submissions

History

Publications

Flanks

ALFA Allele Frequency

The ALFA project provide aggregate allele frequency from dbGaP. More information is available on the project [page](#) including descriptions, data access, and terms of use.

Release Version: 20201027095038

Search:

SNPを調べてみよう

Frequency	Variant Details	Clinical Significance	HGVS	Submissions	History	Publications	Flanks
-----------	-----------------	-----------------------	------	-------------	---------	--------------	--------

Genomic Placements

Sequence name	Change
ALDH2 RefSeqGene	NG_012250.2:g.42076G>A
GRCh37.p13 chr 12	NC_000012.11:g.112241766G>A
GRCh38.p14 chr 12	NC_000012.12:g.111803962G>A

下にスクロールすると、
以下のように表示

Gene: ALDH2, aldehyde dehydrogenase 2 family member (plus strand)

Molecule type	Change	Amino acid[Codon]	SO Term
aldehyde dehydrogenase, mitochondrial isoform 1 precursor	NP_000681.2:p.Glu504Lys	E (Glu) > K (Lys)	Missense Variant
aldehyde dehydrogenase, mitochondrial isoform 2 precursor	NP_001191818.1:p.Glu457Lys	E (Glu) > K (Lys)	Missense Variant
ALDH2 transcript variant 1	NM_000690.4:c.1510G>A	E [GAA] > K [AAA]	Coding Sequence Variant
ALDH2 transcript variant 2	NM_001204889.2:c.1369G>A	E [GAA] > K [AAA]	Coding Sequence Variant

G(グアニン)→A(アデニン)に変異し、それによってアミノ酸がE(グルタミン酸)→K(リジン)に変異

Genomic regions, transcripts, and products

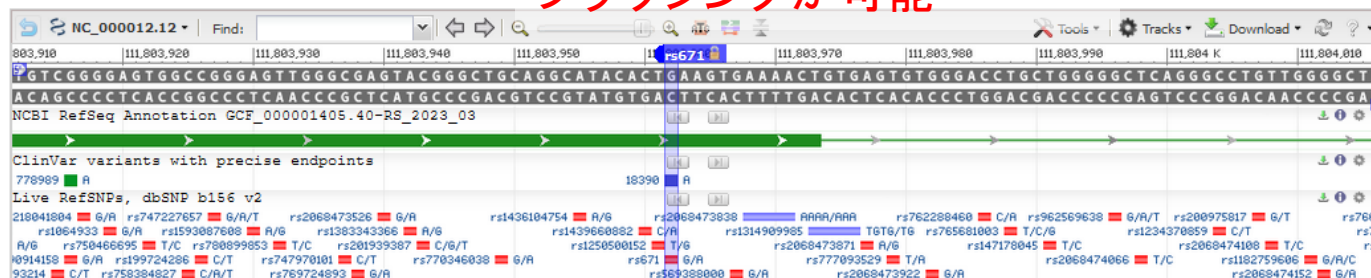
Top ▲ ?

Choose placement

GRCh38.p14 (NC_000012.12) ▾

ゲノム上での位置を示す
ブラウジングが可能

See rs671 in Variation Viewer



コドン表

第1塩基	第2塩基								第3塩基
	U		C		A		G		
U	UUU	Phe (F)	UCU	Ser (S)	UAU	Tyr (Y)	UGU	Cys (S)	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu (L)	UCA		UAA	終止	UGA	終止	A
	UUG		UCG		UAG	終止	UGG	Try (Y)	G
C	CUU	Leu (L)	CCU	Pro (P)	CAU	His (H)	CGU	Arg (R)	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln (Q)	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile (I)	ACU	Thr (T)	AAU	Asn (N)	AGU	Ser (S)	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys (K)	AGA	Arg (R)	A
	AUG	Met (M) 開始	ACG		AAG		AGG		G
G	GUU	Val (V)	GCU	Ala (A)	GAU	Asp (D)	GGU	Gly (G)	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu (E)	GGA		A
	GUG		GCG		GAG		GGG		G

SNPを調べてみよう

Frequency

Variant Details

Clinical Significance

HGVS

Submissions

History

Publications

Flanks

「Clinical Significance」
(臨床上的意味)を選択

Allele: A (allele ID: 33429)

ClinVar Accession	Disease Names	Clinical Significance
RCV000020058.7	Alcohol sensitivity, acute	Drug-Response
RCV000020059.5	Alcohol dependence	Protective
RCV000020060.5	Susceptibility to hangover	Risk-Factor
RCV000020061.5	Sublingual nitroglycerin, susceptibility to poor response to	Risk-Factor
RCV000020062.5	Esophageal cancer, alcohol-related, susceptibility to	Risk-Factor
RCV001290000.2	AMED syndrome, digenic	Pathogenic
RCV001787815.1	ethanol response - Toxicity	Drug-Response

アルコール過敏症(急性)
アルコール依存
二日酔いのなりやすさ
舌下ニトログリセリン
の反応不良に影響
食道がんに影響

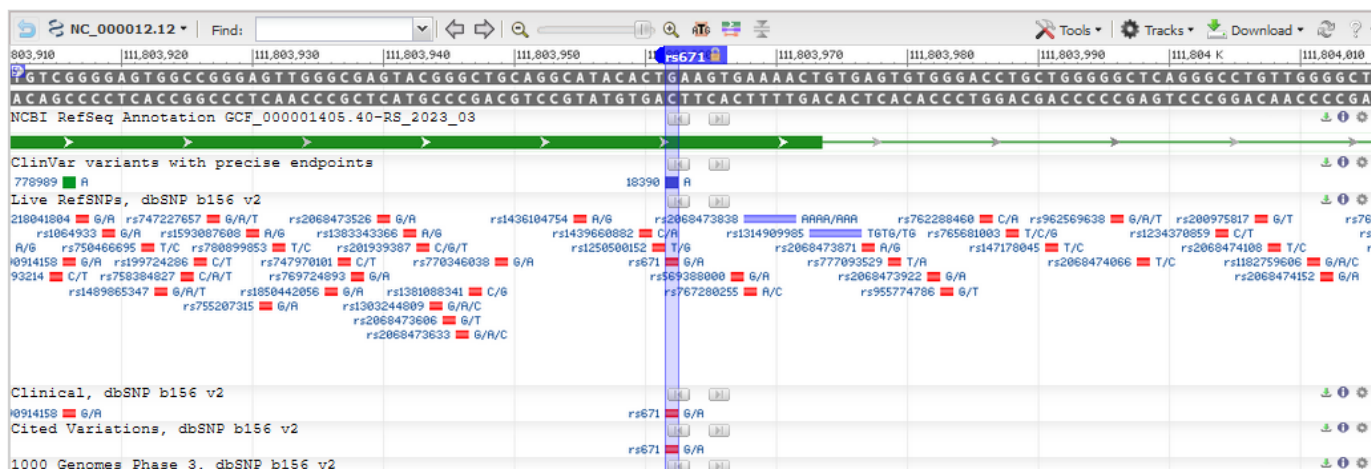
Genomic regions, transcripts, and products

Top ▲ ?

Choose placement

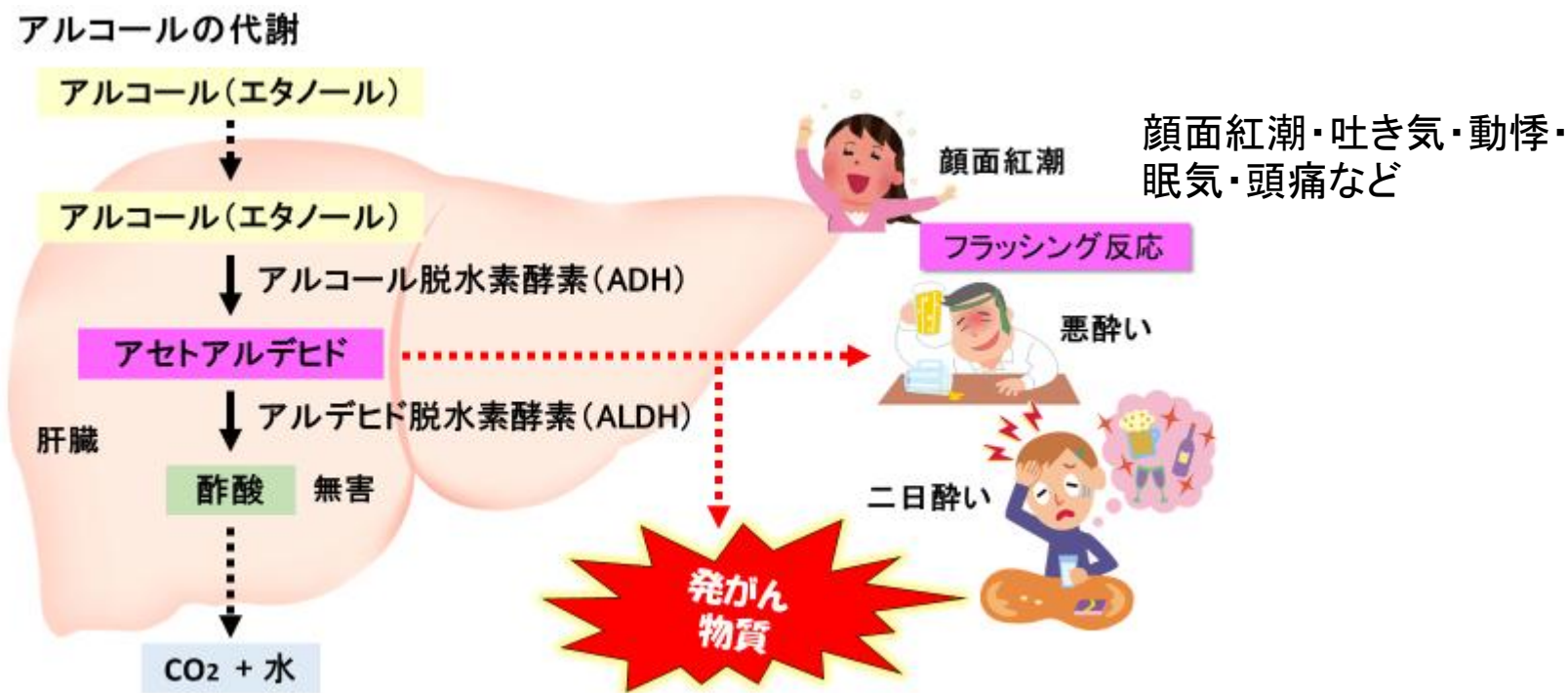
GRCh38.p14 (NC_000012.12) ▾

See rs671 in Variation Viewer



SNPを調べてみよう

「ALDH活性の弱い（酒に弱い）人でよく酒を飲む人の**食道がん**の発生率は、ALDH活性が強い（酒に強い）人で酒をよく飲む人の**12倍**にもなっています（久里浜医療センター 横山顕）」



くにちか内科クリニックのWebページより

2006.03.17

遺伝的に酒に弱い人はニトロ舌下錠が効かない？！ 一酸化窒素放出にアセトアルデヒド脱水素酵素が関与

日本人や中国人に多い生まれつき酒に弱い人では、狭心症の治療に用いるニトログリセリンが効きにくい可能性があることがこのほど明らかになった。アルデヒド脱水素酵素（ALDH2）が、アセトアルデヒドの代謝とニトログリセリンからの一酸化窒素（NO）生成の両方にかかわるため、胎児の肝臓を用いた実験では、酒に弱い遺伝子変異を持つ人では、ニトログリセリンの代謝効率はほぼ10分の1にとどまった。こうした遺伝子を持つかどうかは、アルコールパッチテストや検査機関のテストで容易に分かるため、狭心症治療時にも応用できる可能性がある。中国復旦大学Yifeng Li氏らの研究成果で、詳細はJournal of Clinical Investigation誌2006年2月号に報告された。

ニトログリセリンは、1世紀以上にわたって狭心症や心不全の治療に用いられてきた。ニトログリセリンは、一酸化窒素（NO）の生成を通じて平滑筋を弛緩させる。ニトログリセリンは、代謝されると、NOまたはNO供与体であるS-ニトロソチオールを放出する。NOは、cGMPの活性化を経て平滑筋を弛緩させる。先頃、ニトログリセリンからのNO放出にALDH2が重要な役割を果たすことが明らかになった。飲酒により生じたアセトアルデヒドを酢酸に変える酵素としてよく知られている。

Li氏らは、中国人の狭心症患者80人を調べ、ALDH2にアルコール代謝の能力を大きく減じる変異がある場合、ニトログリセリンに反応しない確率が高くなることを示した。

ALDH2のエクソン12に見出される一般的な変異（504位がグルタミンからリシンに置き換わっている＊）は、アセトアルデヒドを代謝する能力を失わせる。Glu/Lysのヘテロ接合の場合、活性はGluホモ型の約6％、Lysホモ型なら活性は全くない。世界的に見る




医師 人気記事ランキング

	昨日	週間	月間
1	Dr.Kの「医師のためのバリュー投資戦略」 知っているようで知らない「生活保護」の実態		
2	編集会議 on the Web 今週の見逃せない記事（2023年10月9日～20…		

- 薬師寺泰匡の「だから救急はおもしろいよ」
学会シーズン到来！次にバズるフォントはこれだ！…



Association between *ALDH2* and *ADH1B* polymorphisms, alcohol drinking and gastric cancer: a replication and mediation analysis

Kuka Ishioka^{1,2} · Hiroyuki Masaoka^{2,3} · Hidemi Ito^{2,4} · Isao Oze² · Seiji Ito⁵ · Masahiro Tajika⁶ · Yasuhiro Shimizu⁵ · Yasumasa Niwa⁶ · Shigeo Nakamura¹ · Keitaro Matsuo^{2,4} 

ALDH2遺伝子の変異（特にLysアレル）が、飲酒に伴うアセトアルデヒドの影響を介して、胃がんの発症に関与している可能性があることを示唆

Received: 24 January 2018 / Accepted: 15 March 2018 / Published online: 3 April 2018

© The International Gastric Cancer Association and The Japanese Gastric Cancer Association 2018

Abstract

Background Aldehyde dehydrogenase 2 (*ALDH2*; rs671, Glu504Lys) and alcohol dehydrogenase 1B (*ADH1B*; rs1229984, His47Arg) polymorphisms have a strong impact on carcinogenic acetaldehyde accumulation after alcohol drinking. To date, however, evidence for a significant *ALDH2*–alcohol drinking interaction and a mediation effect of *ALDH2/ADH1B* through alcohol drinking on gastric cancer have remained unclear. We conducted two case–control studies to validate the interaction and to estimate the mediation effect on gastric cancer.

Methods We calculated odds ratios (OR) and 95% confidence intervals (CI) for *ALDH2/ADH1B* genotypes and alcohol drinking using conditional logistic regression models after adjustment for potential confounding in the HEPACC-2 (697 cases and 1372 controls) and HEPACC-3 studies (678 cases and 678 controls). We also conducted a mediation analysis of the combination of the two studies to assess whether the effects of these polymorphisms operated through alcohol drinking or through other pathways.

Results *ALDH2* Lys alleles had a higher risk with increased alcohol consumption compared with *ALDH2* Glu/Glu (OR for heavy drinking, 3.57; 95% CI 2.04–6.27; *P* for trend = 0.007), indicating a significant *ALDH2*–alcohol drinking interaction (*P*_{interaction} = 0.024). The mediation analysis indicated a significant positive direct effect (OR 1.67; 95% CI 1.38–2.03) and a protective indirect effect (OR 0.84; 95% CI 0.76–0.92) of the *ALDH2* Lys alleles with the *ALDH2*–alcohol drinking interaction. No significant association of *ADH1B* with gastric cancer was observed.

Conclusion The observed *ALDH2*–alcohol drinking interaction and the direct effect of *ALDH2* Lys alleles may suggest the involvement of acetaldehyde in the development of gastric cancer.

Keywords Alcohol drinking · *ALDH2* · *ADH1B* · Gastric cancer · Interaction · Mediation analysis

SNPを調べてみよう

Frequency

Variant
Details

Clinical
Significance

HGVS

Submissions

History

Publications

Flanks

「Frequency」
(頻度)を選択

ALFA Allele Frequency

The ALFA project provide aggregate allele frequency from dbGaP. More information is available on the project page including descriptions, data access, and terms of use.

Release Version: 20201027095038

ここでは、同じ位置に違う塩基が存在しているものをアレル(allele)と呼ぶ
その頻度を集団ごとに調べた結果

Search:

Population	Group	Sample Size	Ref Allele	Alt Allele
Total	Global	317236	G=0.993945	A=0.006055
European	Sub	273638	G=0.999927	A=0.000073
African	Sub	8512	G=0.9995	A=0.0005
African Others	Sub	304	G=0.997	A=0.003
African American	Sub	8208	G=0.9996	A=0.0004
Asian	Sub	6806	G=0.8008	A=0.1992
East Asian	Sub	4902	G=0.7821	A=0.2179
Other Asian	Sub	1904	G=0.8487	A=0.1513
Latin American 1	Sub	1276	G=1.0000	A=0.0000
Latin American 2	Sub	1862	G=0.9995	A=0.0005
South Asian	Sub	5218	G=0.9990	A=0.0010

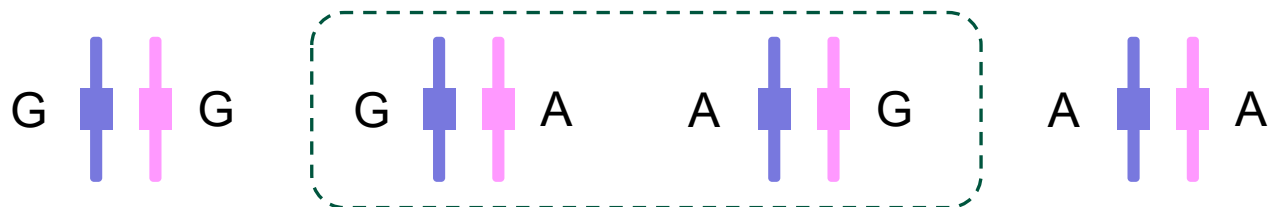
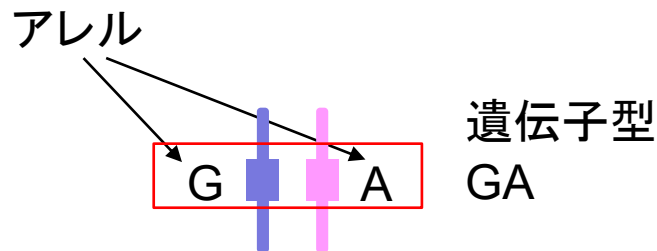
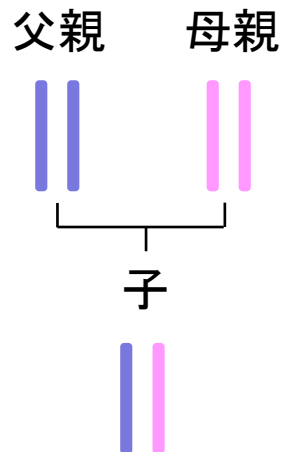
アジア人、とくに東アジア人はもとのG
から変異している人の割合が多い

Download ?

Search:

Study	Population	Group	Sample Size	Ref Allele	Alt Allele
1000Genomes	Global	Study-wide	5008	G=0.9643	A=0.0357
1000Genomes	African	Sub	1322	G=0.9985	A=0.0015
1000Genomes	East Asian	Sub	1008	G=0.8264	A=0.1736
1000Genomes	Europe	Sub	1006	G=1.0000	A=0.0000
1000Genomes	South Asian	Sub	978	G=1.0000	A=0.0000

遺伝子型



片方がG, 片方がA
という点で同じ

遺伝子型
個体が持つ2つのアレルの組み合わせ

GG

GA

AA

タンパク質
(酵素)

機能する

機能が低下する
(機能の低下が大きい)

機能しない

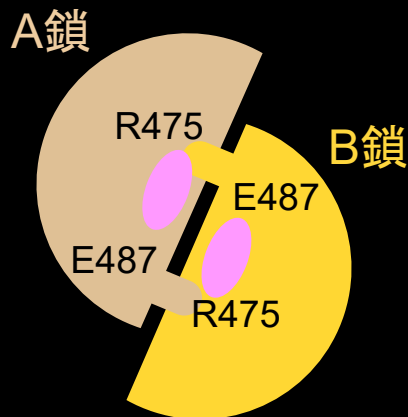
ALDH2の構造

変異なし 3INJ

A鎖 B鎖

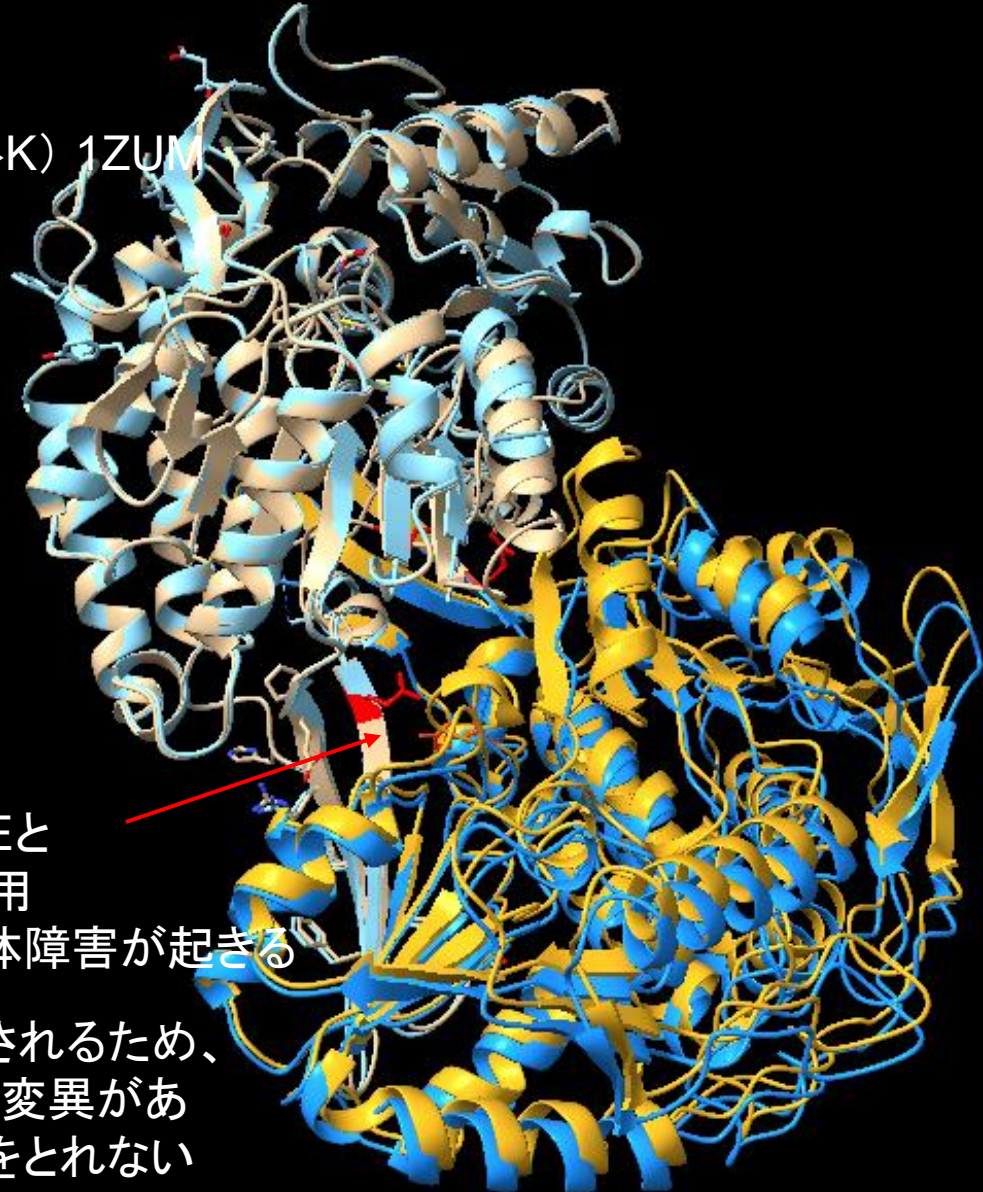
変異あり(487番目のE→K) 1ZUM

A鎖 B鎖



487番目のA鎖のEと
B鎖のRが相互作用
Kに変異すると立体障害が起きる

4つの鎖から形成されるため、
そのうちの1つでも変異があ
ると安定した構造をとれない



SNPの検索

An official website of the United States government [Here's how you know](#) ✓

NIH National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP

Advanced 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](#)

i ALFA Project Release 3 with over 900M variants from >200K subjects is now [available](#) (August 3, 2023)

The goal is to provide allele frequency from more than 1 million dbGaP subjects with regular updates. Visit the project [page](#) for more information or view the introduction video below.

 ALFA CoLab Presentation, ASHG2020

後で見る 共有

NCBI ALFA

「ADH1B」と入力して
「Search」ボタンをクリック

SNPの検索

An official website of the United States government [Here's how you know](#)

NIH National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP

SNP Search

Create alert Advanced Help

Clinical Significance
benign
likely benign
protective

Validation Status
by-ALFA
by-cluster
by-frequency

Publication
PubMed Cited
PubMed Linked

Function Class
inframe deletion
initiator codon variant
intron
missense
synonymous

Variation Class
del
delins
ins
mnv

Annotation
somatic

Global MAF
Custom range...

[Clear all](#)
[Show additional filters](#)

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

Send to: Filters: Manage Filters

Find related data
Database: Select Find items

Search details
ADH1B[All Fields]
Search See more...

Recent activity
Turn Off Clear

ADH1B (7158) SNP
ALH (0) SNP
ALH1B (0) SNP
Homo sapiens aldehyde dehydrogenase 2 family member (ALDH2), transcr Nucleotide
See more...

Search results
Items: 1 to 20 of 7158

<< First < Prev Page 1 of 358 Next > Last >>

☒ rs1229984 [Homo sapiens]

1. Variant type: SNV
Alleles: T>A,C,G [Show Flanks]
Chromosome: 4:99318162 (GRCh38)
4:100239319 (GRCh37)
Canonical SPDI: NC_000004.12:99318161:T:A,NC_000004.12:99318161:T:C,NC_000004.12:99318161:T:G
Gene: ADH1B (Varview)
Functional Consequence: missense_variant,coding_sequence_variant
Clinical significance: protective
Validated: by frequency,by alfa,by cluster
MAF: T=0.057792/2524 (ALFA)
T=0.0 (GENOME_DK)
T=0.0 (PRJEB37584)
HGVS: NC_000004.12:g.99318162T>A, NC_000004.12:g.99318162T>C,
NC_000004.12:g.99318162T>G, NC_000004.11:g.100239319T>A,
NC_000004.11:g.100239319T>C, NC_000004.11:g.100239319T>G,
NC_000004.11:g.100239319T>A, NC_000004.11:g.100239319T>C, NC_000004.11:g.100239319T>G,
NC_000004.11:g.100239319T>A, NC_000004.11:g.100239319T>C, NC_000004.11:g.100239319T>G, ...more
[PubMed](#) [LitVar](#)

☐ rs2018417 [Homo sapiens]

2. Variant type: SNV
Alleles: C>A,T [Show Flanks]
Chromosome: 4:99313983 (GRCh38)
4:100235140 (GRCh37)
Canonical SPDI: NC_000004.12:99313982:C:A,NC_000004.12:99313982:C:T
Gene: ADH1B (Varview)
Functional Consequence: missense_variant,coding_sequence_variant

多数のSNPの中から、先頭の「rs1229984」を選択

SNPを調べてみよう

dbSNP Short Genetic Variations

[Search](#)

Examples: rs268, BRCA1 and more

[Advanced search](#)

Welcome to the Reference SNP (rs) Report

All alleles are reported in the [Forward orientation](#). Click on the [Variant Details](#) tab for details on Genomic Placement, Gene, and Amino Acid changes. HGVS names are in the [HGVS](#) tab.

Reference SNP (rs) Report

参照配列では
T(チミン)→A(アデニン)、C(シ
トシン)、G(グアニン)に変異

[Download](#)

Current Build 157

Released September 3, 2024

rs1229984

Organism

Homo sapiens

Position

chr4:99318162 (GRCh38.p14) [?](#)

Alleles

T>A / T>C / T>G

Variation Type

SNV Single Nucleotide Variation

Frequency

T=0.068824 (18217/264690, TOPMED)
T=0.093623 (11366/121402, ExAC)
C=0.24662 (19099/77444, 38KJPN) (+ 22 more)

Clinical Significance

Reported in [ClinVar](#)

Gene : Consequence

ADH1B : Missense Variant

Publications

252 citations

[LitVar²](#) 839

Genomic View

[See rs on genome](#)

Frequency

Variant
Details

Clinical
Significance

HGVS

Submissions

History

Publications

Flanks

ALFA Allele Frequency

The ALFA project provide aggregate allele frequency from dbGaP. More information is available on the project [page](#) including descriptions, data access, and terms of use.

Release Version: 20250407153717

Search:

Feedback

SNPを調べてみよう

Frequency	Variant Details	Clinical Significance	HGVS	Submissions	History	Publications	Flanks
Genomic Placements							
Sequence name	Change						
ADH1B RefSeqGene	NG_011435.1:g.8254A>T						
ADH1B RefSeqGene	NG_011435.1:g.8254A>G						
ADH1B RefSeqGene	NG_011435.1:g.8254A>C						
GRCh37.p13 chr 4	NC_000004.11:g.100239319T>A						
GRCh37.p13 chr 4	NC_000004.11:g.100239319T>C						
GRCh37.p13 chr 4	NC_000004.11:g.100239319T>G						
GRCh38.p14 chr 4	NC_000004.12:g.99318162T>A						
GRCh38.p14 chr 4	NC_000004.12:g.99318162T>C						
GRCh38.p14 chr 4	NC_000004.12:g.99318162T>G						
LOC126807122 genomic region	NG_083443.1:g.559T>A						
LOC126807122 genomic region	NG_083443.1:g.559T>C						
LOC126807122 genomic region	NG_083443.1:g.559T>G						
Gene: ADH1B, alcohol dehydrogenase 1B (class I), beta polypeptide (minus strand)							
Molecule type	Change	Amino acid[Codon]	SO Term				
ADH1B transcript variant 1	NM_000668.6:c.143A>T	H [CAC] > L [CTC]	Coding Sequence Variant				
ADH1B transcript variant 1	NM_000668.6:c.143A>G	H [CAC] > R [CGC]	Coding Sequence Variant				
ADH1B transcript variant 1	NM_000668.6:c.143A>C	H [CAC] > P [CCC]	Coding Sequence Variant				
ADH1B transcript variant 2	NM_001286650.2:c.23A>T	H [CAC] > L [CTC]	Coding Sequence Variant				
ADH1B transcript variant 2	NM_001286650.2:c.23A>G	H [CAC] > R [CGC]	Coding Sequence Variant				

-鎖なので、AとT, CとGは置き換えて解釈する

下にスクロールする

—鎖なので、AとT, CとGは
置き換えて解釈する

下にスクロールすると…

コドン表

第1塩基	第2塩基								第3塩基
	U		C		A		G		
U	UUU	Phe (F)	UCU	Ser (S)	UAU	Tyr (Y)	UGU	Cys (S)	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu (L)	UCA		UAA	終止	UGA	終止	A
	UUG		UCG		UAG	終止	UGG	Try (Y)	G
C	CUU	Leu (L)	CCU	Pro (P)	CAU	His (H)	CGU	Arg (R)	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln (Q)	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile (I)	ACU	Thr (T)	AAU	Asn (N)	AGU	Ser (S)	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys (K)	AGA	Arg (R)	A
	AUG	Met (M) 開始	ACG		AAG		AGG		G
G	GUU	Val (V)	GCU	Ala (A)	GAU	Asp (D)	GGU	Gly (G)	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu (E)	GGA		A
	GUG		GCG		GAG		GGG		G

SNPを調べてみよう

Gene: **ADH1B**, alcohol dehydrogenase 1B (class I), beta polypeptide (minus strand)

Molecule type	Change	Amino acid[Codon]	SO Term
ADH1B transcript variant 1	NM_000668.6:c.143A>T	H [CAC] > L [CTC]	Coding Sequence Variant
ADH1B transcript variant 1	NM_000668.6:c.143A>G	H [CAC] > R [CGC]	Coding Sequence Variant
ADH1B transcript variant 1	NM_000668.6:c.143A>C	H [CAC] > P [CCC]	Coding Sequence Variant
ADH1B transcript variant 2	NM_001286650.2:c.23A>T	H [CAC] > L [CTC]	Coding Sequence Variant
ADH1B transcript variant 2	NM_001286650.2:c.23A>G	H [CAC] > R [CGC]	Coding Sequence Variant
ADH1B transcript variant 2	NM_001286650.2:c.23A>C	H [CAC] > P [CCC]	Coding Sequence Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 1	NP_000659.2:p.His48Leu	H (His) > L (Leu)	Missense Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 1	NP_000659.2:p.His48Arg	H (His) > R (Arg)	Missense Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 1	NP_000659.2:p.His48Pro	H (His) > P (Pro)	Missense Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 2	NP_001273579.1:p.His8Leu	H (His) > L (Leu)	Missense Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 2	NP_001273579.1:p.His8Arg	H (His) > R (Arg)	Missense Variant
all-trans-retinol dehydrogenase [NAD(+)] ADH1B isoform 2	NP_001273579.1:p.His8Pro	H (His) > P (Pro)	Missense Variant

アミノ酸がH(ヒスチジン)→
R(アルギニン)に変異

変異はすべて
失活となる

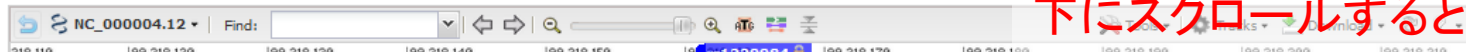
Genomic regions, transcripts, and products

Top ▲ ⓘ

Choose placement

GRCh38.p14 (NC_000004.12)

See rs1229984 in Variation Viewer



下にスクロールすると…

SNPを調べてみよう

Genomic regions, transcripts, and products

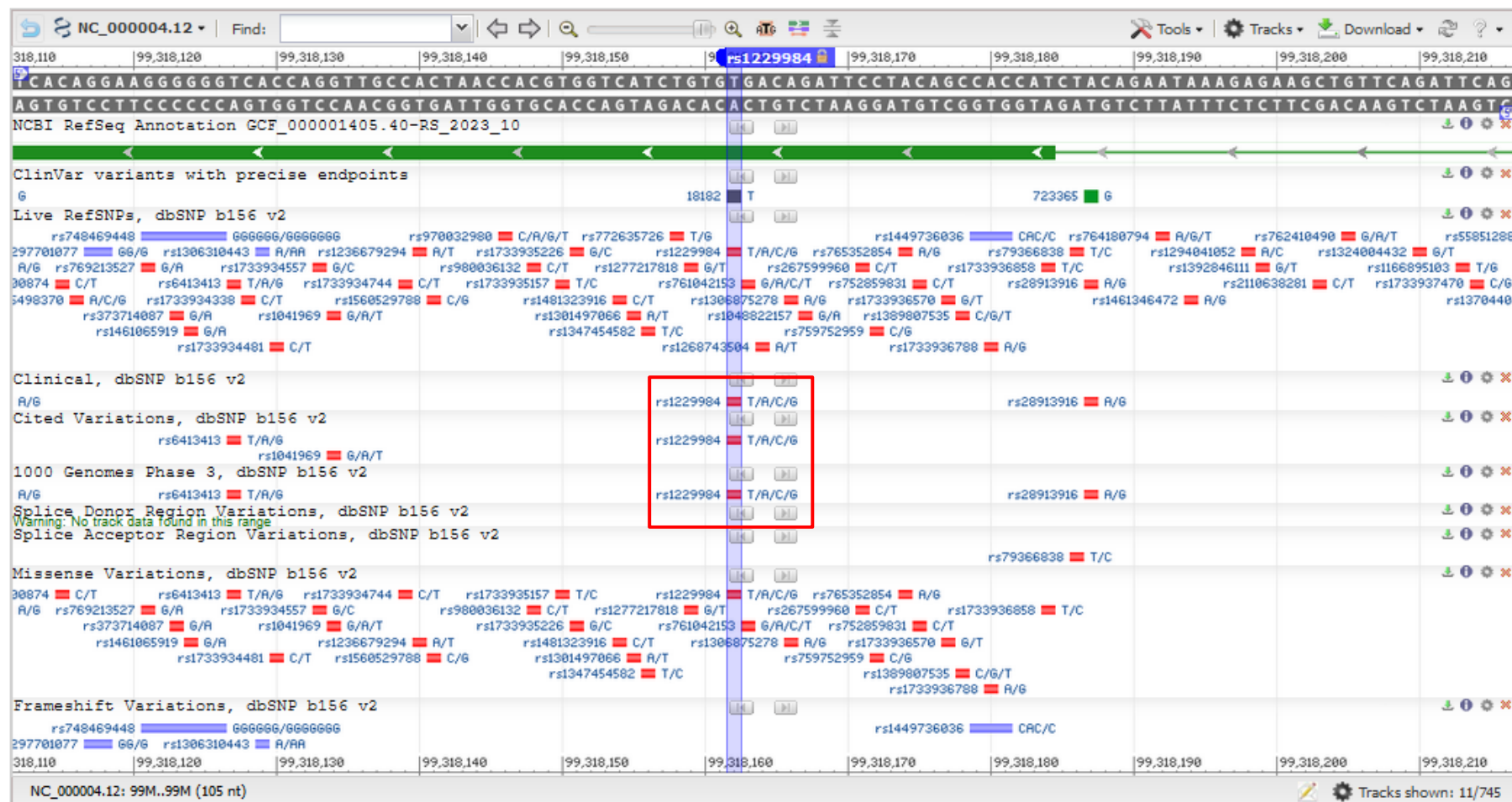
ゲノム上での位置を示す
ブラウジングが可能

Top ▲ ?

Choose placement

GRCh38.p14 (NC_000004.12) ▼

See rs1229984 in Variation Viewer



SNPを調べてみよう

Frequency

Variant
Details

Clinical
Significance

HGVS

Submissions

History

Publications

Flanks

「Clinical Significance」
(臨床上的意味)を選択

Allele: T= (allele ID: 33221)

ClinVar Accession	Disease Names	Clinical Significance
RCV000019813.5	Alcohol dependence	Protective
RCV000019814.5	Aerodigestive tract cancer, squamous cell, alcohol-related, protection against	Protective

疾患のリスク
を減少させる

Genomic regions, transcripts, and products

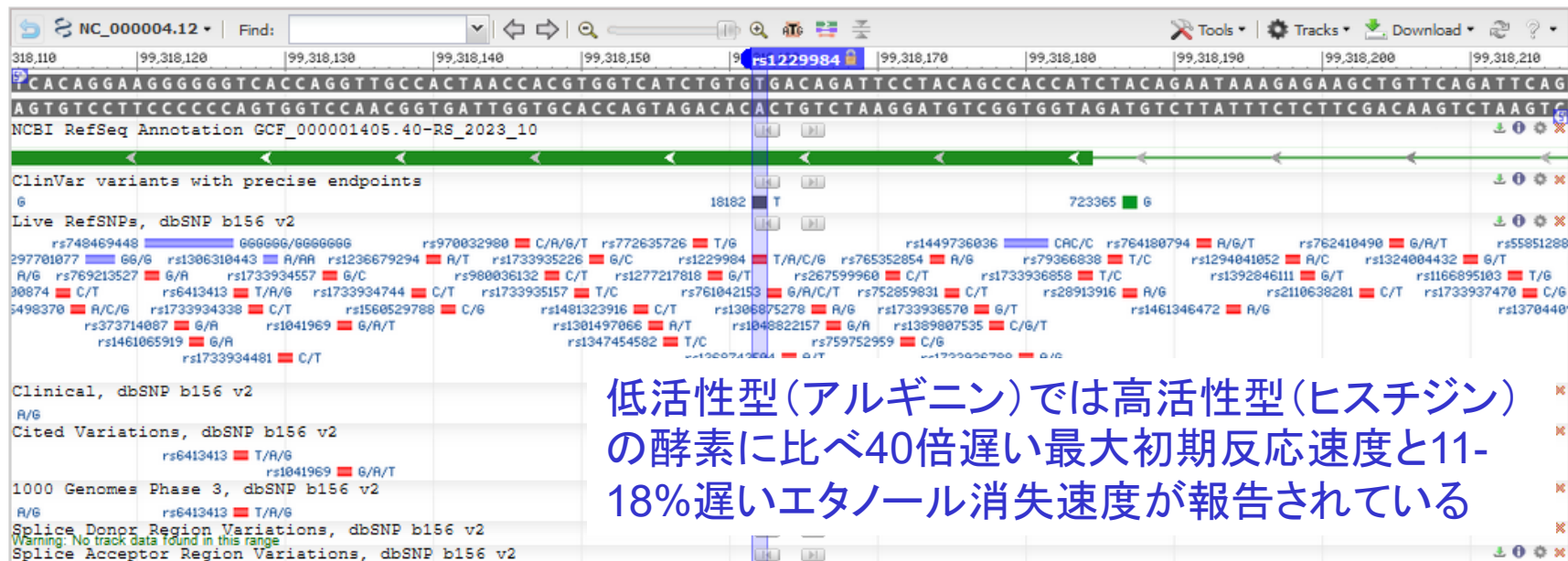
Top ▲ ?

Choose placement

GRCh38.p14 (NC_000004.12)

See rs1229984 in Variation Viewer

Tアレルを持っていると、
アルコール関連の上部消
化がんにかかりにくい



低活性型(アルギニン)では高活性型(ヒスチジン)の酵素に比べ40倍遅い最大初期反応速度と11-18%遅いエタノール消失速度が報告されている

SNPを調べてみよう

Frequency

Variant
Details

Clinical
Significance

HGVS

Submissions

History

Publications

Flanks

「Frequency」 「Allele Frequency」

(頻度)を選択

The ALFA project provides aggregate allele frequency from dbGaP. More information is available on the project [page](#) including descriptions, data access, and terms of use.

Release Version: 20230706150541

Search:

Population	Group	Sample Size	Ref Allele	Alt Allele
Total	Global	59934	T=0.04882	C=0.95118, G=0.00000
European	Sub	49190	T=0.04462	C=0.95538, G=0.00000
African	Sub	6004	T=0.0238	C=0.9762, G=0.0000
African Others	Sub	198	T=0.010	C=0.990, G=0.000
African American	Sub	5806	T=0.0243	C=0.9757, G=0.0000
Asian	Sub	162	T=0.827	C=0.173, G=0.000
East Asian	Sub	118	T=0.814	C=0.186, G=0.000
Other Asian	Sub	44	T=0.86	C=0.14, G=0.00
Latin American 1	Sub	6	T=1.0	C=0.0, G=0.0
Latin American 2	Sub	20	T=0.00	C=1.00, G=0.00
South Asian	Sub	6	T=0.2	C=0.8, G=0.0

アジア人はほと
のAの人の割合
が多い

一鎖なので、Cは
G、TはAを表す

A
ヒスチジン


G
アルギニン

[Download](#) [?](#)

Search:

Study	Population	Group	Sample Size	Ref Allele	Alt Allele
-------	------------	-------	-------------	------------	------------

SNPの検索

**National Library of Medicine**
National Center for Biotechnology Information

Log in

dbSNP

SNP

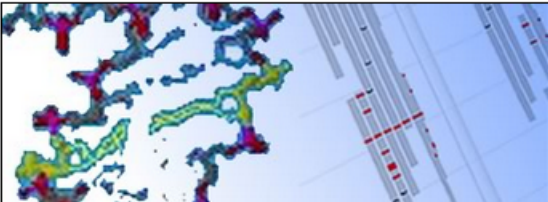
SLC24A5

Search

Advanced

Help

**「SLC24A5」と入力して
「Search」ボタンをクリック**

**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](#)

**ALFA Project Release 3 with over 900M variants from >200K subjects is now [available](#) (August 3, 2023)**

The goal is to provide allele frequency from more than 1 million dbGaP subjects with regular updates. Visit the project [page](#) for more information or view the introduction video below.



ALFA CoLab Presentation, ASHG2020

後で見る 共有

米国の国立医療機関によるもの

NCBI ALFA

SNPの検索



National Library of Medicine
National Center for Biotechnology Information

Log in

dbSNP

SNP

SLC24A5



Search

Create alert Advanced

Help

Clinical
Significance
benign
conflicting interpretations of
pathogenicity
likely benign
likely pathogenic
pathogenic

Validation Status
by-ALFA
by-cluster
by-frequency

Publication
PubMed Cited
PubMed Linked

Function Class
inframe deletion
inframe indel
inframe insertion
initiator codon variant
intron
missense
synonymous

Variation Class
del
delins
ins

Annotation
somatic

Global MAF
Custom range...

Clear all
Show additional filters

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

Send to: Filters: Manage Filters

Search results

Items: 1 to 20 of 8664

<< First < Prev Page 1 of 434 Next > Last >>

☒ rs1426654 [Homo sapiens]

1. Variant type: SNV
Alleles: A>G,T [Show Flanks]
Chromosome: 15:48134287 (GRCh38)
15:48426484 (GRCh37)
Canonical SPDI: NC_000015.10:48134286:A:G,NC_000015.10:48134286:A:T
Gene: MYEF2 (Varview), SLC24A5 (Varview)
Functional Consequence: downstream_transcript_variant,500B_downstream_variant,5_prime_UTR_variant,coding_sequence_var
Clinical significance: association,benign
Validated: by frequency,by alfa,by cluster
MAF: G=0.068875/21666 (ALFA)
G=0./0 (PRJEB36033)
G=0.002157/8 (TWINSUK)

...more

HGVS: NC_000015.10:g.48134287A>G, NC_000015.10:g.48134287A>T,
NC_000015.9:g.48426484A>G, NC_000015.9:g.48426484A>T,
NG_011500.1:g.18316A>G, NG_011500.1:g.18316A>T, NM_205850.3:c.331A>G,
NM_205850.3:c.331A>T, NM_205850.3:c.331A>C, NM_205850.3:c.331A>T

...more

PubMed LitVar

☐ rs76547866 [Homo sapiens]

2. Variant type: SNV
Alleles: A>T [Show Flanks]
Chromosome: 15:48141109 (GRCh38)
15:48433306 (GRCh37)
Canonical SPDI: NC_000015.10:48141108:A:T
Gene: MYEF2 (Varview), SLC24A5 (Varview)

多数のSNPの中から、先頭の「rs1426654」を選択

Find related data

Database: Select

Find items

Search details

SLC24A5[A11 Fields]

Search

See more...

Recent activity

Turn Off Clear

SLC24A5 (8664)

SNP

ALDH2 (18788)

SNP

See more...

血液型を決める遺伝子

- ABO遺伝子（ABO糖転移酵素を作る遺伝子）
- SNPによって酵素の機能が異なる
→ 血液型の違いにつながる

血液型を決める遺伝子を調べてみよう

Genome Data Viewer

GDV supports the exploration and analysis of [NCBI-annotated](#) and selected non-NCBI annotated eukaryotic genome assemblies. Currently, assemblies from over 2290 organisms are available.

Switch view

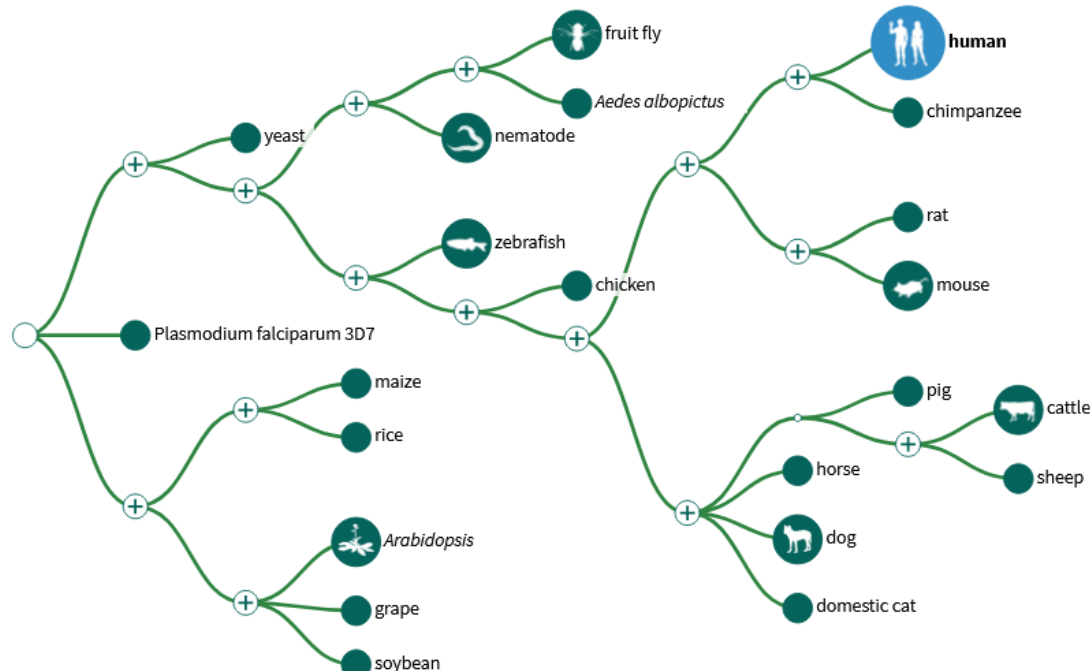


Search organisms

Homo sapiens (human)

To view more organisms in the tree, click on nodes that have '+' signs. Press and hold the '+' to expand and reveal all the subgroups.
Or, search for an organism using the search box above.

New! Click on Switch view at the top to see another way of navigating genomes.



ABOと入力

Homo sapiens (human)



Search in genome

ABO



Examples: TP53, chr17:7667000-7689000, DNA repair

Assembly

GRCh38.p14

Browse genome

Compare genomes

...

Assembly details

Name GRCh38.p14
RefSeq accession [GCF_000001405.40](#)
GenBank accession [GCA_000001405.29](#)
Submitter Genome Reference Consortium
Level Chromosome
Category Reference genome

Annotation details

Annotation Release RS_2023_03 [i](#)
Release date Mar 20, 2023



ABO遺伝子のブラウジング

Genome Data Viewer

Homo sapiens
(human)

Assembly: GRCh38.p14 (GCF_000001405.40)

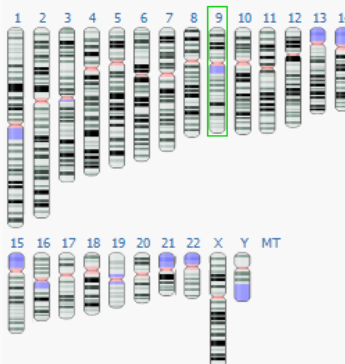
Chr 9 (NC_000009.12)

Home Share this page Reset All More Tools More Info

Search assembly
Q ABO
Examples

Assemblies
Ideogram View

Unplaced/unlocalized scaffolds: 166
Alt loci/patches: 514



Tracks and User Data
BLAST
Tracks by Accession
Assembly Region Details
History

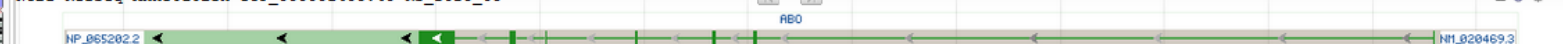
NC_000009.12: 133,247,921 - 133,277,681



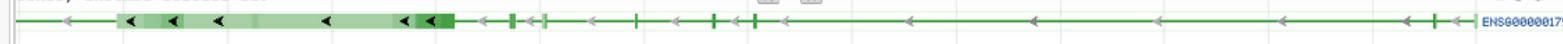
Region ABO Transcript NM_020469.3 Exons: click an exon to zoom in, mouse over to see details

NC_000009.12 133,248 K 133,250 K 133,252 K 133,254 K 133,256 K 133,258 K 133,260 K 133,262 K 133,264 K 133,266 K 133,268 K 133,270 K 133,272 K 133,274 K 133,276 K

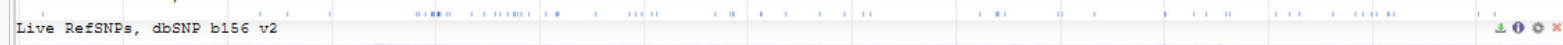
NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03



Genes, Ensembl release 110



Cited Variations, dbSNP b156 v2



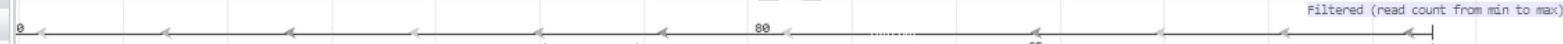
Live RefSNPs, dbSNP b156 v2



RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled



RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled



RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation...



NC_000009.12: 133M..133M (29,761 nt)

Tracks shown: 8/942

ABO遺伝子のブラウジング

Genome Data Viewer

Homo sapiens
(human)

Assembly: GRCh38.p14 (GCF_000001405.40)

Chr 9 (NC_000009.12)

Home Share this page Reset All More Tools More Info

Search assembly

ABO

Examples

Assemblies

Ideogram View

Unplaced/unlocalized scaffolds:
Alt loci/patches:

166
514



Tracks and User Data

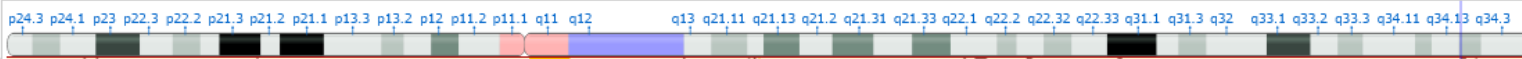
BLAST

Tracks by Accession

Assembly Region Details

History

NC_000009.12: 133,247,921 - 133,277,681



Region ABO Transcript NM_020469.3 Exons: click an exon to zoom in, mouse over to see details

NC_000009.12 133,248 K 133,250 K 133,252 K 133,254 K 133,256 K 133,258 K 133,260 K 133,262 K 133,264 K 133,266 K 133,268 K 133,270 K 133,272 K 133,274 K 133,276 K

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

NP_065202.2 ABO NM_020469.3

Genes, Ensembl release 110

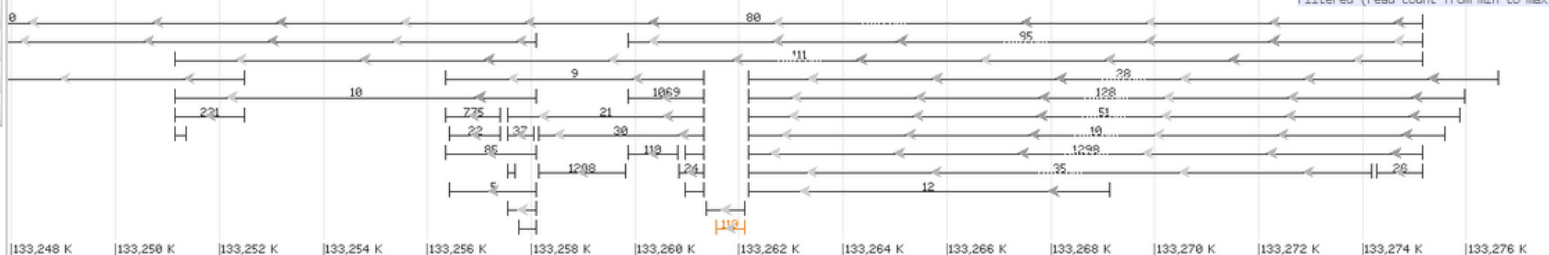
Cited Variations, dbSNP b156 v2

Live RefSNPs, dbSNP b156 v2

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled



Cited Variations, dbSNP
b156 v2の設定マークを指定

ABO遺伝子のブラウジング

Genome Data Viewer

Homo sapiens
(human)

Assembly: GRCh38.p14 (GCF_000001405.40)

Chr 9 (NC_000009.12)

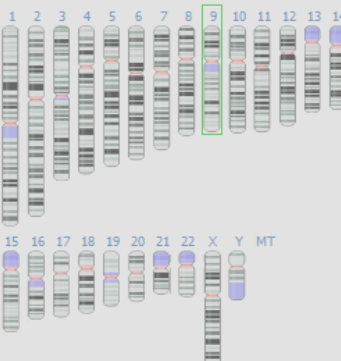
Home Share this page Reset All More Tools More Info

Search assembly
Q ABO
Examples

Assemblies

Ideogram View

Unplaced/unlocalized scaffolds: 166
Alt loci/patches: 514



Tracks and User Data

BLAST

Tracks by Accession

Assembly Region Details

History

NC_000009.12: 133,247,921 - 133,277,681

p24.3 p24.1 p23 p22.3 p22.2 p21.3 p21.2 p21.1 p13.3 p13.2 p11.2 p11.1 q11 q12 q13 q21.11 q21.13 q21.2 q21.31 q21.33 q22.1 q22.2 q22.32 q22.33 q31.1 q31.3 q32 q33.1 q33.2 q33.3 q34.11 q34.13 q34.3

Region ABO Transcript NM_020469.3 Exons: click an exon to zoom in, mouse over to see details

NC_000009.12 133,248 K 133,250 K 133,252 K 133,254 K 133,256 K 133,258 K 133,260 K 133,262 K 133,264 K 133,266 K 133,268 K 133,270 K 133,272 K 133,274 K 133,276 K

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

NP_065202.2 ABO NT_020469.3

Genes, Ensembl release 110

Cited Variations, dbSNP b156 v2

Live RefSNPs, dbSNP b156 v2

RNA-seq exon coverage, aggregate (filtered), base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation...

Filtered (read count from min to max)

8 10 9 1069 128 51 10 1298 12 28

221 775 22 32 30 119 24 142

5 1208 12

133,248 K 133,250 K 133,252 K 133,254 K 133,256 K 133,258 K 133,260 K 133,262 K 133,264 K 133,266 K 133,268 K 133,270 K 133,272 K 133,274 K 133,276 K

NC_000009.12: 133M..133M (29,761 nt)

Tracks shown: 8/942

Cited Variations, dbSNP b156 v2

Rendering options:

- Show variants for 50 or less
- Show variants for 50 or less
- Collapse on single line
- Show all
- Show variants with labels

Track legend

「Show variants with labels」
を指定

ABO遺伝子のブラウジング

Genome Data Viewer

Homo sapiens
(human)

Assembly: GRCh38.p14 (GCF_000001405.40)

Chr 9 (NC_000009.12)

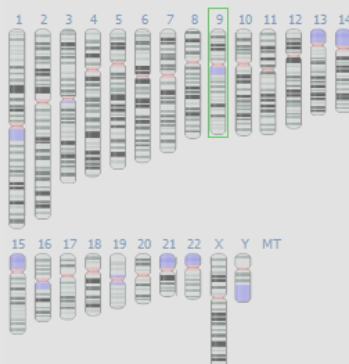
Home Share this page Reset All More Tools More Info

Search assembly
Q ABO
Examples

Assemblies

Ideogram View

Unplaced/unlocalized scaffolds: 166
Alt loci/patches: 514



Tracks and User Data

BLAST

Tracks by Accession

Assembly Region Details

History

NC_000009.12: 133,247,921 - 133,277,681

p24.3 p24.1 p23 p22.3 p22.2 p21.3 p21.2 p21.1 p13.3 p13.2 p12 p11.2 p11.1 q11 q12 q13 q21.11 q21.13 q21.2 q21.31 q21.33 q22.1 q22.2 q22.32 q22.33 q31.1 q31.3 q32 q33.1 q33.2 q33.3 q34.11 q34.13 q34.3

Region ABO Transcript NM_020469.3 Exons: click an exon to zoom in, mouse over to see details

133,248 K 133,250 K 133,252 K 133,254 K 133,256 K 133,258 K 133,260 K 133,262 K 133,264 K 133,266 K 133,268 K 133,270 K 133,272 K 133,274 K 133,276 K

NCBI RefSeq Annotation GCF_000001405.40-RS_2023_03

HP_065202.2 ABO NT_020469.3 ENSG0000010171

Genes, Ensembl release 110

Cited Variations, dbSNP b156 v2

Rendering options: Show variants with labels

Track legend

Apply

Cancel

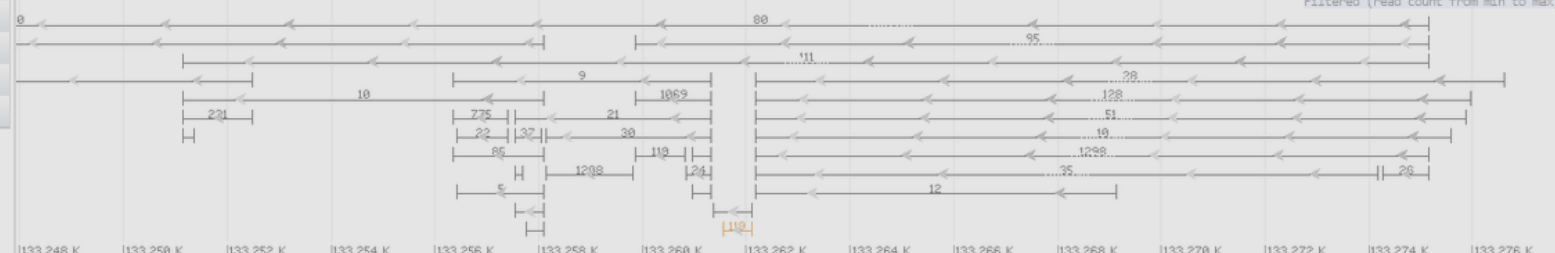
「Apply」ボタンをクリック

Live RefSNPs, dbSNP b156 v2

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation...



NC_000009.12: 133M..133M (29,761 nt)

Tracks shown: 8/942

NIH National Library of Medicine
National Center for Biotechnology Information

Genome Data Viewer

[Home](#)
[Share this page](#)
[Reset All](#)
[More Tools▼](#)
[More Info▼](#)

Assembly: GRCh38.p14 (GCF_000001405.40) ▾ • Chr 9 (NC_000009.12) ▾

NC_000009.12: 133,247,921 - 133,277,681

9p21.2

p24.3 p24.1 p23 p22.3 p22.2 p21.3 p21.2 p21.1 p11.2 p11.1 p11 p10 p10.2 p10.1 p10 p9.3 p9.2 p9.1 p9 p8.3 p8.2 p8.1 p8 p7.3 p7.2 p7.1 p7 p6.3 p6.2 p6.1 p6 p5.3 p5.2 p5.1 p5 p4.3 p4.2 p4.1 p4 p3.3 p3.2 p3.1 p3 p2.3 p2.2 p2.1 p2 p1.3 p1.2 p1.1 p1 p0.3 p0.2 p0.1 p0

q13 q21.11 q21.13 q21.2 q21.31 q21.33 q22.1 q22.2 q22.32 q22.33 q31.1 q31.3 q32 q33.1 q33.2 q33.3 q34.11 q34.13 q34.3

Region Gene Transcript Exons: click an exon to zoom in, mouse over to see details

NC_000009.12  Tools  Tracks  Download  ?



Genes, Ensembl release 110

Cited Variations. dbSNP b156 v2

269 | R/G rs10901252 | G/R/C/T rs56392308 | GGG/G/G/G/6666 rs638758 | A/R/C/G rs680976 | C/A rs8176685 | CGCCACCCACTACGCC... rs545971 | T/R/C rs2769071 | G/A rs659104 | G/R/T/rs532436 | A/G/T rs552
rs7857390 | A/C/G/G/T rs8176719 | -/C rs8176707 | G/T rs687621 | G/R/C rs657152 | A/C/T rs2519693 | T/C rs8176668 | A/C/T rs676457 | T/R/C rs500498 | C/R/T rs568203 | C/R/G rs
rs11244053 | R/G rs8176747 | C/R/G rs512778 | G/A/C/T rs687289 | A/C/G rs474279 | C/R/G/T rs8176672 | C/G/T rs28850884 | A/C rs660340 | G/R/C/T rs600038 | C/T
rs8176751 | C/R/G/T rs641959 | A/C/T rs2073828 | A/R/C rs8176681 | T/C rs514659 | C/G/T rs644234 | G/R/T rs643434 | A/G/T rs550057 | T/R/C rs8176645 | A/G/T
rs8176749 | C/T rs8176717 | G/T rs549446 | C/T rs8176694 | T/C rs8176693 | C/T rs672316 | T/R/C/G rs612169 | G/A rs505922 | C/T
rs41382905 | C/G/T rs641943 | A/C/G/T rs8176745 | G/R/rs8176714 | G/A rs624601 | G/R/C rs8176704 | G/A rs507666 | A/G
rs8176746 | G/R/T rs514708 | C/T rs574347 | T/C rs8176743 | C/G/T rs55722397 | G/C rs8176742 | C/T rs8176741 | G/A
rs8176740 | A/R/C/T rs7853969 | G/R/C rs1053878 | G/A rs8176734 | C/T rs8176732 | A/G/T rs2073823 | G/A
rs2073824 | A/G rs8176722 | C/A rs8176720 | T/R/C/G rs25129845 | T/C rs25129845 | T/C
rs1053878 | G/A

Live RefSNPs. dbSNP b156 v2

RNA-seq exon coverage, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron-spanning reads, aggregate (filtered), NCBI Homo sapiens Annotation Release 110 - log base 2 scaled

RNA-seq intron features, aggregate (filtered), NCBI Homo sapiens Annotation...


Filtered (read count from min to max)


37

血液型の決まり方

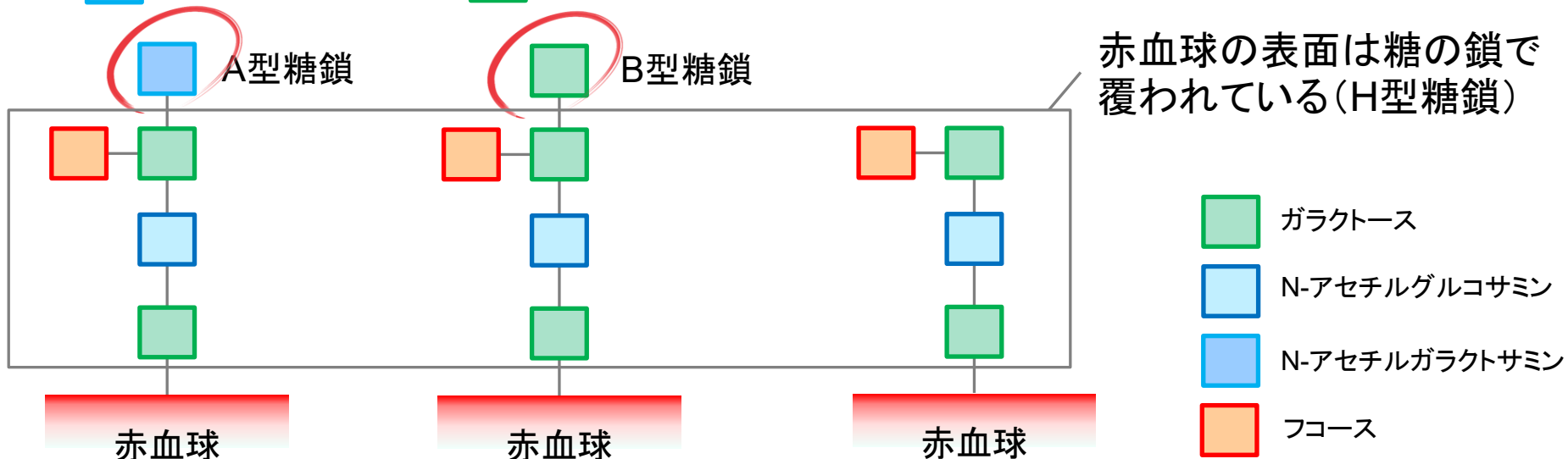
ゴルジ体で働く糖転移酵素の遺伝子（ABO遺伝子）の主なSNP

SNP id	遺伝子の配列	タンパク質の配列	遺伝子
変異なし（A型の遺伝子を基準にした場合）			A型
rs8176746	796番目のC→A	266番目のL→M	B型
rs8176747	803番目のG→C	268番目のG→A	
rs8176743	703番目のG→A	235番目のG→S	
rs8176719	261番目のGの欠失	3文字単位がずれて短いタンパク質になり、酵素の機能を失う	O型

A型遺伝子が作る酵素
 を付加（転移）

B型遺伝子が作る酵素
 を付加

O型遺伝子は糖を付加しない



血液型の決まり方

父親 母親



子



父と母からの遺伝子の組み合わせ

遺伝子型

AA

AO

BB

BO

AB

OO

血液型

A型

B型

AB型

O型

A型糖鎖
(持たない
B, O型の
人にとって
A抗原)

B型糖鎖
(持たない
A, O型の
人にとって
B抗原)

A型糖鎖
(A抗原) B型糖鎖
(B抗原)

A型抗体 ×
B型抗体 ○

A型抗体 ○
B型抗体 ×

A型抗体 ×
B型抗体 ×

A型抗体 ○
B型抗体 ○

赤血球

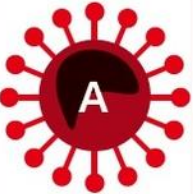
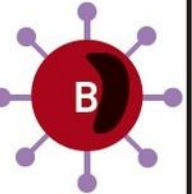
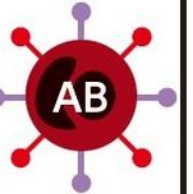
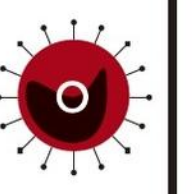



赤血球

赤血球

赤血球

血液型の決まり方

【抗原・抗体対照表】 日本赤十字社

	A 型	B 型	AB 型	O 型
赤血球型				
抗原 (赤血球)	A 抗原	B 抗原	A・B 抗原	抗原なし
抗体 (血漿)			抗体なし	
	抗 B 抗体	抗 A 抗体	抗体なし	抗 A・抗 B 抗体

- ▶ 赤血球を輸血する場合は抗原を合わせる(抗原のないO型は万能)
- ▶ 血漿を輸血する場合は抗体を合わせる(抗体のないAB型は万能)

- ・ 抗原: 生体に免疫応答を引き起こす物質
- ・ 抗体: 抗原を体外へ排除するために作られるタンパク質(免疫グロブリン)

赤血球の輸血

		輸血者			
		A	B	AB	O
供血者	A	○	×	○	×
	B	×	○	○	×
	AB	×	×	○	×
	O	○	○	○	○

血漿の輸血

		輸血者			
		A	B	AB	O
供血者	A	○	×	×	○
	B	×	○	×	○
	AB	○	○	○	○
	O	×	×	×	○

実際の全血の輸血では、血漿中の抗体の問題があり、同型のみ輸血が行われる

SNPedia

- SNPedia

- DNAバリエーションに関する情報を文献情報をもとにまとめたサイト

- <https://www.snpedia.com/>

SNPedia

[Talk](#) [? Contributions](#) [Create account](#) [Log in](#)

Search SNPedia



Navigation ▾

[Main page](#) [Discussion](#) [Edit](#) [History](#)

Have questions? Visit <https://www.reddit.com/r/SNPedia>



SNPedia

SNPedia is a wiki investigating human genetics. We share information about the effects of variations in DNA, citing peer-reviewed scientific publications. It is used by [Promethease](#) to create a personal report linking your DNA variations to the information published about them. Please see the [SNPedia:FAQ](#) for answers to common questions.

Help! [\[edit\]](#)

- look at the example [rs1234](#)
- learn more about SNPs

SNPediaの利用

SNPedia

Talk ? Contributions Create account Log in

blood type



「blood type」と入力



Search results

Q blood type



Search

Results 1 – 22 of 358

Content pages Multimedia Everything Advanced

Create the page "**Blood type**" on this wiki! See also the search results found.

Page title matches

ABO Blood Type

#REDIRECT [[ABO blood group]]

29 bytes (4 words) - 18:23, 27 July 2014

「ABO blood Type」をクリック

Page text matches

Rs7903146

}}This [[SNP]] in [[TCF7L2]] influences the risk of [[Type-2 diabetes]] ([[T2D]]). This SNP is also known as IVS3C>T. }} [[rs7903146(C;T)]] [[rs7903146(T;T)]] strongly predicted future [[type-2 diabetes]]. Considered in context with [[rs7903146]]

SNPediaの利用

ABO blood group

(Redirected from [ABO Blood Type](#))

The **ABO blood group** system is the best known blood typing system, determining blood group type as A, B, AB, or O, and thereby of importance for blood donors and recipients. [Wikipedia](#) Three common alleles (A, B and O) of the [ABO](#) gene yield the six common combinations that a person can have, each of them resulting in a particular ABO blood type (as determined serologically):

Alleles	ABO Type
A,A	A
A,B	AB
A,O	A
B,B	B
B,O	B
O,O	O

SNPs in the [ABO](#) gene determine the alleles present and thereby predict ABO blood group type.

[23andMe](#) checks these 11 snps

- [rs8176719](#)
- [rs1053878](#)
- [rs7853989](#)
- [rs8176740](#)
- [rs8176743](#)
- [rs8176746](#)
- [rs41302905](#)
- [rs8176747](#)
- [i4000504](#)
- [rs8176749](#)
- [i4000505](#)

However, determination of blood group type can be via [genosets](#) is shown in [this spreadsheet](#) or this more complicated and comprehensive one

- [rs8176719](#)

課題 2

SLC24A5遺伝子のSNP rs1426654について調べてみよう。

1. A（アデニン）がG（グアニン）に変異することにより、この遺伝子がコードするタンパク質の何番目のアミノ酸が何から何に変異するか？また、この変異によって活性はどのように変化するか？
2. この変異は、どのような地域に多く見られるか？
3. この変異は、人の外見のどのようなことに関係するか？

SLC24A5は、ヒト表皮におけるメラニン新生を制御する、K依存性Na-Ca交換活性を持つタンパク質である。この遺伝子の変異により交換活性が変化し、メラニン生成が抑制されることで、皮膚の色が白くなり、紫外線の少ない高緯度地域でのビタミンD合成が有利になった。その結果、進化の過程でヨーロッパ人集団において広く普及したと考えられる。

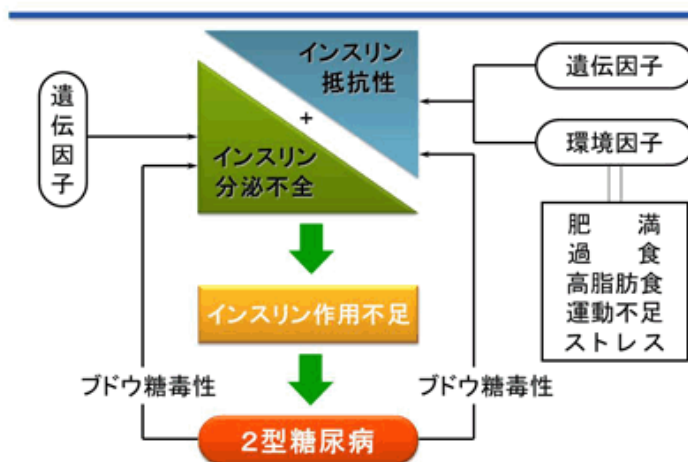
課題 3

SNPediaを用いて、2型糖尿病（Type-2 diabetes）に関する遺伝子を調べ、その遺伝子の機能およびそのSNPの1つについて調べてみよう。

2型糖尿病とは

最も多いタイプの糖尿病で、一般的に"糖尿病"と表現した場合2型糖尿病を示す事が多いです。遺伝的素因によるインスリン分泌能の低下に、環境的素因としての生活習慣の悪化に伴うインスリン抵抗性が加わり、インスリンの相対的不足に陥った場合に発症する糖尿病です（図1）。一般的に生活習慣病と称されるタイプの糖尿病が2型糖尿病ですが、インスリン分泌能の低下が不可欠です。ですから生活習慣の乱れだけではなく、2型糖尿病患者さんは大なり小なり糖尿病になりやすい体質（遺伝的素因）を持っているとも言えます。

2型糖尿病の病態・成因



日本内分泌学会

http://www.j-endo.jp/modules/patient/index.php?content_id=93

課題 3

Type-2 diabetes

The wikipedia article on [Diabetes mellitus type 2](#) is a good place to start.

In the news (2019): 23andMe's Polygenic Risk Score for type-2 diabetes. A fine, short summary concluding that it is a good "engagement tool" but otherwise of no clinical use can be found [here](#).

[omim](#) summarizes the latest research.

Numerous SNPs have each been associated with (slightly) increased risk for **type-2 diabetes**, but they only marginally improve the odds of predicting whether an individual will get **type-2 diabetes** based on the traditional clinical characteristics combining age, sex and weight ([PMID 18694974]; see also [1]). Such SNPs include:

- 10 SNPs in all 10 regions found so far in multiple studies and meta-analyses, as known at the time of publication [PMID 17463248]:
 - rs4402960, in the IGF2BP2 gene region, equivalent to rs1470579
 - rs7754840, in CDKAL1 gene, equivalent to rs10946398 and rs4712523
 - rs10811661, in an intergenic region on ch 9 near CDKN2A
 - rs9300039, in an intergenic region on ch11, and 96% of time equivalent to rs1514823
 - rs8050136, in the FTO gene region
 - rs1801282 and rs17036314, in the PPARC gene
 - rs13266634, in the SLC30A8 gene
 - rs1111875 and rs7923837, near the HHEX gene, and equivalent to rs5015480
 - rs7903146, in the TCF7L2 gene, and 85% of the time equivalent to rs7901695
 - rs5219, in the KCNJ11 gene, and 99% of the time equivalent to rs5215
 - Note that this paper reports that if you have the highest risk genotype for all 10 of these SNPs, you are estimated to be at double the risk for **type-2 diabetes** compared to the average person, whereas if you have the lowest risk genotype for every one of these 10 SNPs, you are estimated to be at half the risk.
- The 12 SNPs reported in a (2007) large, multi-lab Consortium study [PMID 17554300]:
 - rs9465871, in the CDKAL1 gene fifth intron, like rs775840 and others
 - rs4506565, in the TCF7L2 gene, and 92% of the time equivalent to rs7903146
 - rs9939609, in the FTO gene, like rs9939609, rs7193144 and rs8050136
 - rs4655595

例えば、SLC30A8遺伝子のSNPは？