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Japanese Multi Omics  
Reference Panel.

全ゲノムリファレンスパネル

# 4.7KJPNデータ利用ガイド



## ゲノムバリエーション



## Genome Variation

統計情報  
(4.7KJPN)

コホート名	人数
東北メディカル・メガバンク計画による宮城県と岩手県でのコホート調査への協力者	4,378 (うち993人が新規追加)
独立行政法人国立病院機構長崎医療センターにおける協力者	188
ながはま0次予防コホート事業における協力者	40
国立がん研究センターにおける協力者	47 (新規追加)
J-MICC Studyにおける協力者	60 (新規追加)
大阪大学眼科における協力者	30 (新規追加)
大阪大学ツインリサーチセンターにおける協力者	30 (新規追加)
合計	4,773

Category	# of total SNVs	# of total INDELS
Autosome	59,199,967	8,238,823
chrX (PAR1+PAR2)	2,316,891	341,289
chrX(PAR1+PAR2+XTR)	2,318,572	341,547
chrMT	2,801	



ゲノムバリエント



Genome Variation

## トップページ

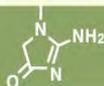


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Phenome

To be provided



Metabolome



Proteome



Transcriptome

Iwate Medical Megabank Organization; iMethyl



Methylome

Iwate Medical Megabank Organization; iMethyl



Genome Variation



Genome Sequence

jMorp release 201911

November, 1st, 2019

GWAS summary statistics datasets and Japonica Array NEO marker list

We have added GWAS summary statistics page, which is a list of Genome-Wide Association Studies performed by TMM project. Users can download summary statistics data for

**代謝物 15K** Array NEO is available from Downloads page.

[More](#)

**タンパク質 500**

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

Tadaka S, Katsuoka F, et al.,  
"3.5KJPNv2, An allele frequency panel of 3,552 Japanese Individuals including the X chromosome"  
Human Genome Variation, 2019 Jun 18;6:28. doi: 10.1038/s41439-019-0059-5. [↗](#)

Saigusa D, Matsukawa N, Tadaka S, Motoike IN, Koshiba S.  
"Metabolome Analysis of Human Plasma by GC-MS/MS in a Large-scale Cohort"  
Proteome Letters, 2019 Volume 4 Issue 1 Pages 31-40. doi: 10.14889/jpros.4.1\_31. [↗](#)

[More](#)

**ゲノムバリエント 4.7K**

**ゲノム配列**





ゲノムバリエント



Genome Variation

## バリエントを検索



## Genomic Variants

Statistics

Downloads

Search by gene name

Search by rs#

Search by region (GRCh37/hg19)

Gene symbol

Examples: ALDH2, NFE2L2, GATA1

Search

遺伝子名検索

ID検索

染色体上の位置検索





# ゲノムバリエント



# Genome Variation

## 結果一覧

Sequence | Variation | Proteome | Metabolome | Repository | GWAS | Downloads | Help | Login

by gene: **ALDH2** chrCh37hg19

SNV/ INDEL 座標 dbSNP\_ID 機能 遺伝子名 疾患DB 頻度

**Japonica Array**

Type	Position	Ref/Alt	rs#	Annotation	Gene	MeanDepth (162PE)	JPA	ClinVar Annotation	ToMMo 4.7K.JPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ	gnomAD EAS	gnomAD NFE
SNV	12:112237599	C/T	rs965904639	intron_variant	ALDH2	23.0/23.0								0.0001
SNV	12:112237603	G/A		intron_variant	ALDH2	22.0/22.0			0.0001					
SNV	12:112237618	C/T	rs149377733	intron_variant	ALDH2	22.6/22.6				0.0163				0.0001
SNV	12:112237629	A/C	rs184287363	intron_variant	ALDH2	22.0/22.0			0.0002					
SNV	12:112237634	C/T	rs1370369478	intron_variant	ALDH2	22.9/22.9								0.0001
INDEL	12:112237779	TCT	rs1156259069	frameshift_variant (p.Thr441fs)	ALDH2	23.3/23.3							0.0006	
SNV	12:112237783	C/T	rs771107373	missense_variant (p.Thr441Met)	ALDH2	23.0/23.0			0.0003					

バリエントのQuality:  
FILTER=PASS

バリエントのQuality:  
FILTER=not PASS

○略称  
 AFR: African/African American  
 AMR: Admixed American  
 ASJ: Ashkenazi Jewish  
 EAS: East Asian  
 NFE: Non-Finnish European

INDEL	12:112201185	T/TA	rs1301926051	upstream_gene_variant	ALDH2	18.0/18.0								
SNV	12:112201186	A/C	rs1319486146	upstream_gene_variant	ALDH2	18.2/18.1								0.0001
SNV	12:112201187	C/A	rs866155334	upstream_gene_variant	ALDH2	18.0/18.0			0.0023	0.0289	0.0302	0.0244	0.0182	0.0338



## ゲノムバリエント



## Genome Variation

## 結果一覧→詳細表示

「Browser」をクリックしてブラウザを表示

Search by gene: ALDH2 GRCh37/hg19

3963 variants found

Filter by keyword

Type	Position	Ref/Alt	rs#	Annotation	Gene	MeanDepth (162PE)	JPA	ClinVar Annotation	ToMMo 4.7KJPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ	gnomAD EAS	gnomAD NFE
SNV	12:112237599	C/T	rs965904639	Intron_variant	ALDH2	23.0/23.0								0.0001
SNV	12:112237603	G/A		Intron_variant	ALDH2	22.0/22.0			0.0001					
SNV	12:112237618	C/T	rs149377733	Intron_variant	ALDH2	22.6/22.6				0.0163				0.0001
SNV	12:112237629	A/C	rs184287363	Intron_variant	ALDH2	22.0/22.0			0.0002					
SNV	12:112237634	C/T	rs1370369478	Intron_variant	ALDH2	22.9/22.9								0.0001
INDEL	12:112237779	TC/T	rs1156259069	frameshift_variant (p.Thr441fs)	ALDH2	23.3/23.3							0.0005	
SNV	12:112237783	C/T	rs771107373	missense_variant (p.Thr441Met)	ALDH2	23.0/23.0			0.0003					

「Plot」をクリックして集団間の頻度比較を表示

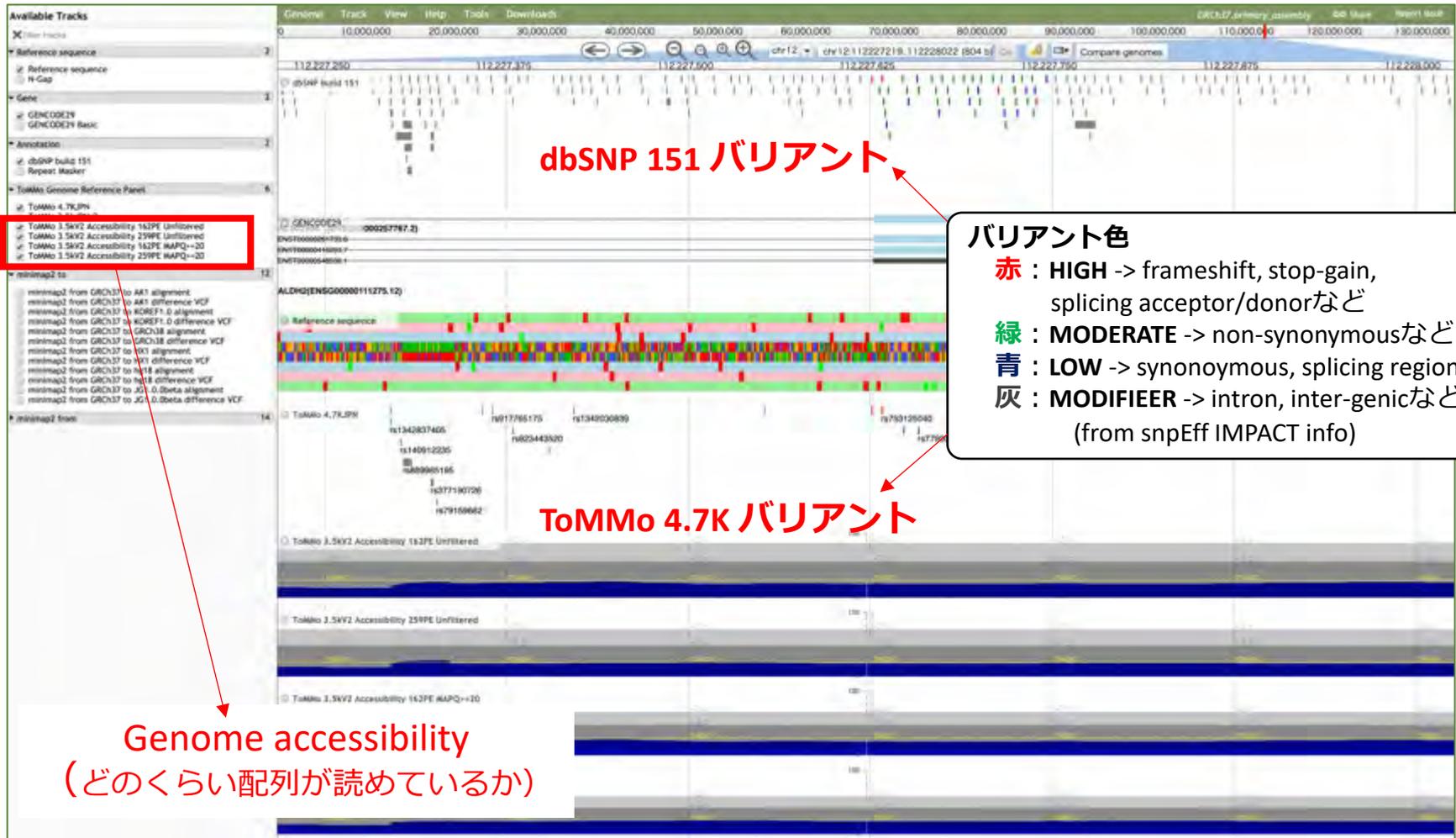


# ゲノムバリエント



# Genome Variation

## 結果一覧→ブラウザ表示



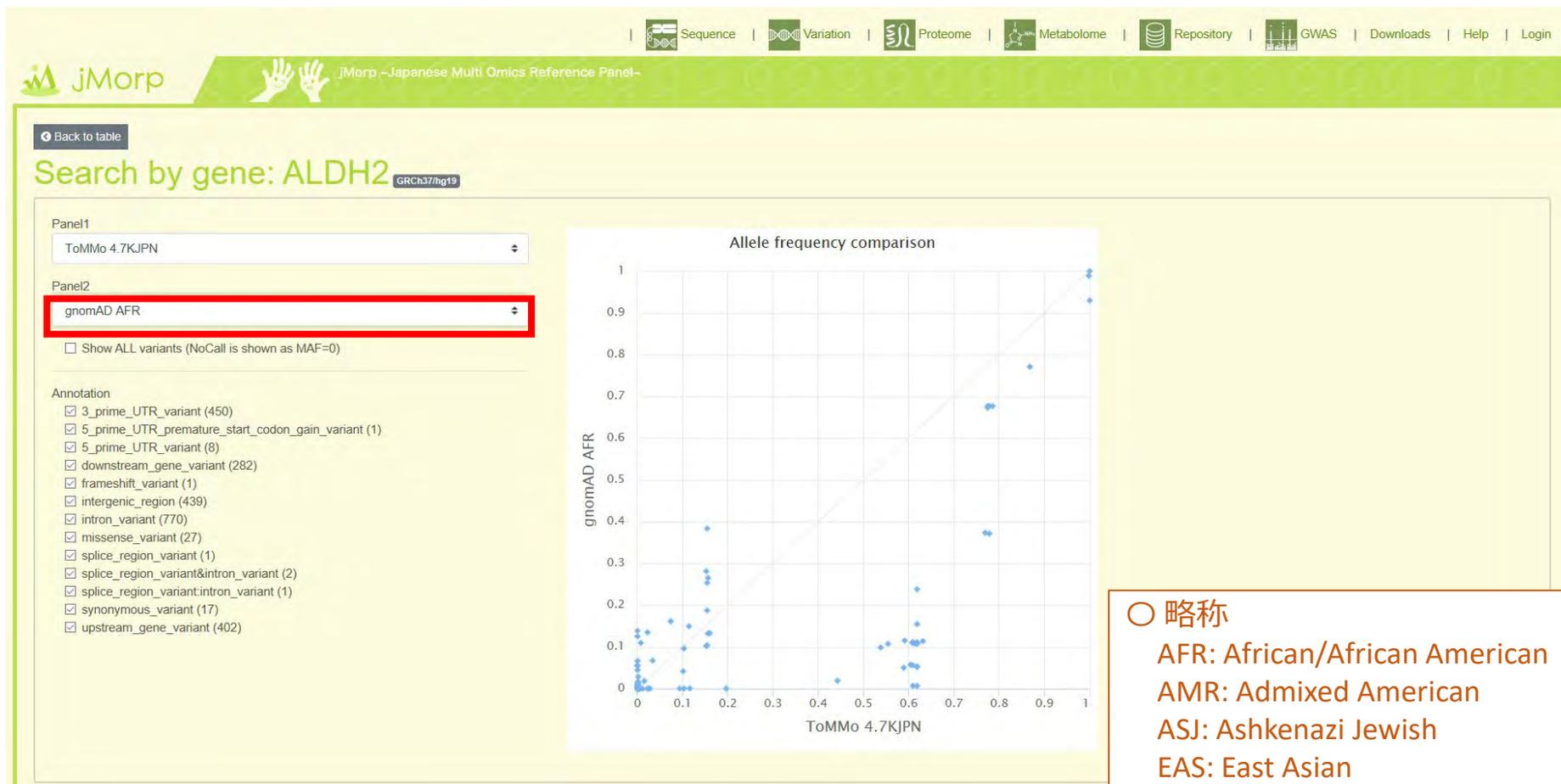


## ゲノムバリエーション



## Genome Variation

## 結果一覧→集団間の頻度比較



## ゲノムバリエント



## Genome Variation

## 結果一覧→タンパク構造へのマッピング



Search by gene: ALDH2

GRCh37/hg19

クリックして表示

47 variants found

Statistics

Browser

Plot

Type	Position	Ref/Alt	rs#	Annotation	Gene	MeanDepth (162PE)	JPA	ClinVar Annotation	ToMMo 4.7KJPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ	gnomAD EAS	gnomAD NFE
SNV	12:112241766	G/A	rs671	missense_variant (p.Glu504Lys)	ALDH2	18.0/18.0	V1&V2&NEO	drug_response	0.1976	0.0002	0.0012		0.2671	
SNV	12:112241742	G/A	rs769724893	missense_variant (p.Glu496Lys)	ALDH2	19.4/19.4								0.0001
SNV	12:112241689	C/G	rs1319756154	missense_variant (p.Ala478Gly)	ALDH2	23.8/23.8							0.0006	
SNV	12:112241677	A/G	rs1454299460	missense_variant (p.Asp474Gly)	ALDH2	24.2/24.2								0.0001
SNV	12:112237855	C/T	rs757893333	missense_variant (p.Ala465Val)	ALDH2	23.0/23.0			0.0001					
SNV	12:112237831	A/G	rs764713289	missense_variant (p.Asn457Ser)	ALDH2	23.6/23.6								0.0001
SNV	12:112237824	A/G	rs776526118	missense_variant (p.Lys455Glu)	ALDH2	23.4/23.4								0.0001
SNV	12:112237783	C/T	rs771107373	missense_variant (p.Thr441Met)	ALDH2	23.0/23.0			0.0003					
SNV	12:112237750	T/C	rs374377652	missense_variant (p.Ile430Thr)	ALDH2	23.5/23.5								0.0001
SNV	12:112236021	A/G	rs1174749347	missense_variant (p.Asp408Gly)	ALDH2	23.7/23.7								0.0001
SNV	12:112236014	G/A	rs1431308860	missense_variant (p.Val406Met)	ALDH2	23.7/23.7				0.0001				



## ゲノムバリエント



## Genome Variation

## 結果一覧→タンパク構造へのマッピング

## 12:112241766 G/A - Structure Mapping

BlastHit1 BlastHit2 BlastHit3 BlastHit4 BlastHit5 BlastHit6 BlastHit7 BlastHit8 BlastHit9 BlastHit10

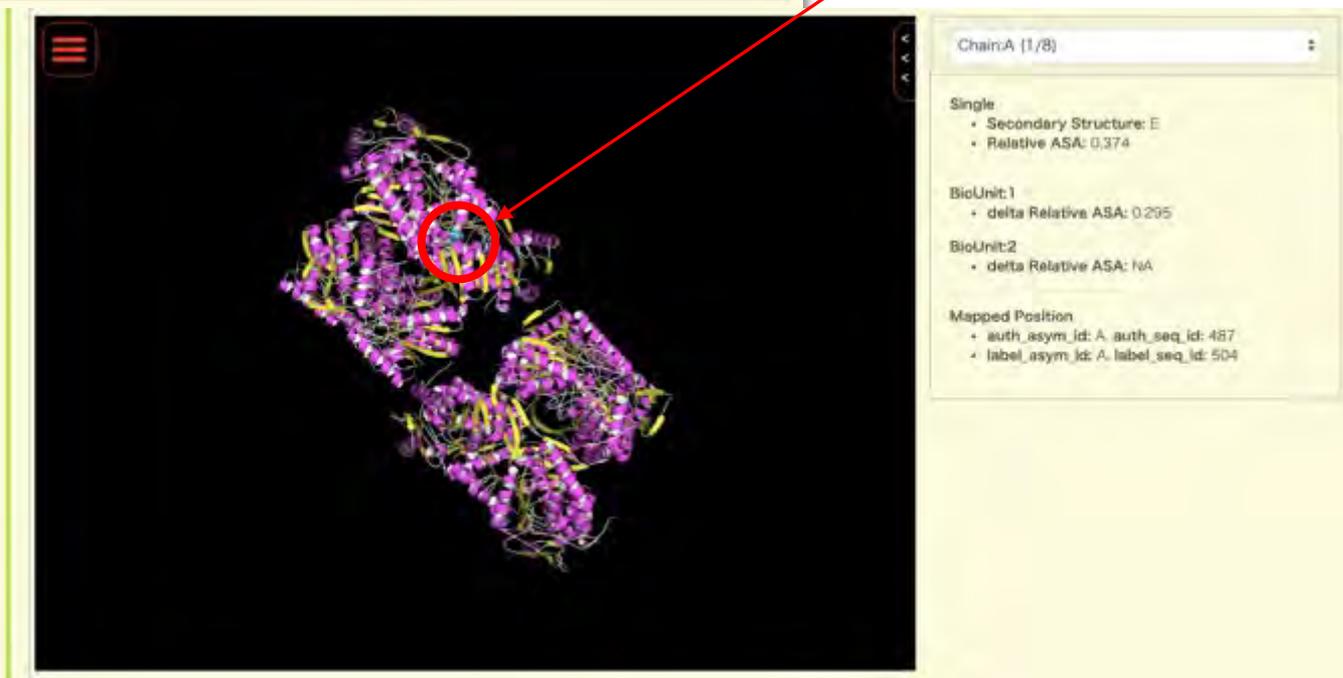
## Blast result

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) [C]   NM_000690.3 [C]   NP_000681.2 [C]
Subject	51132
e-value	0.0000
Sequence Identity	100.00

1510番目の塩基G→A,

504番目のアミノ酸E(グルタミン酸)→K(リシン)

標的バリエント (水色の球)



interactiveに操作可能