



Welcome to
Japanese Multi Omics
Reference Panel.

全ゲノムリファレンスパネル

8.3KJPNデータ利用ガイド

ゲノムバリエント



Genome Variation

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	7,958
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	191
ながはま 0 次予防コホート事業における協力者	65
国立がん研究センターにおける協力者	47
J-MICC Studyにおける協力者	60
大阪大学眼科における協力者	30
大阪大学ツインリサーチセンターにおける協力者	29
合計	8,380

Category	# of total SNVs	# of total INDELs
Autosome	76,324,188	10,017,023
chrX (PAR1+PAR2)	2,994,485	411,478
chrX(PAR1+PAR2+XTR)	3,031,683	415,785
chrMT	3,357	



ゲノムバリエント



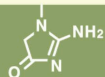
Genome Variation

トップページ

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 202008

August 31st, 2020

代謝物

タンパク質

ゲノムバリエント 8.3K

ゲノム配列

[Reference Genome, 8.3KJPN allele frequency panel, and 25K Metabolome] We updated the Japanese reference genome, JG2, which is constructed by integrating the assemblies from three Japanese male individuals.

[Genome variation] 8.3KJPN, allele and genotype frequency panels from 8,380 Japanese individuals were released.

distributions of concentrations of 45 metabolites identified by NMR detected in the peak intensities of 169 characterized metabolites by GC-MS in 2,939 volunteers. We also released the quantified metabolome data from 2,374 volunteers by new Biocrates LC-MS analysis. Moreover, information of metabolome data obtained from volunteers in repeat assessment survey by GC-MS is released.

The normalization and quality filter protocol was updated from 2019 version in GC-MS data. Three metabolite's name (TCO500085, TCO500226, and TCZ001256) were corrected.

[More](#)

Takahara G, Sugawara D, Motomura H, Imoto S, Hori T, Shimotohno M, Koshida S, Yamamoto M, Kinoshita K. "jMorp: Japanese Multi Omics Reference Panel" *BMC Genomics* 2018 Jan 4;19(1):1-11. doi: 10.1186/s12864-018-4651-5. [PMID: 29211111](#)

"8.3KJPNv2, 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. [PMID: 31111111](#)

ゲノムバリエント



Genome Variation

バリエントを検索



Sequence



Variation



Methylome



Transcriptome



Proteome



Metabolome



Repository



GWAS

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Genomic Variants (8.3KJPN)

[Statistics](#) [Downloads](#)

Search by gene name

Search by rs#

Search by region (GRCh37/hg19)

Gene symbol

ALDH2

Candidates: ALDH2

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 | Methyome | Transcriptome | Proteome | Metabolome



Search by gene: ALDH2 (8.3KJPN)

GRCh37/hg19

Japonica
Array搭載の有無
(v1 / v2 / NEO)

Statistics

Browser Plot

SNV/
INDEL 座標 塩基 dbSNP_ID 機能 遺伝子 深度 疾患DB

集団毎のアレル頻度

Type	Position ↑↓	Ref/Alt	rs#	Annotation ↑↓	Gene ↑↓	MeanDepth (162PE)	JPA ↑↓	ClinVar Annotation	ToMMo 8.3KJPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ
SNV	12:112241766	G/A	rs671	missense_variant (p.Glu504Lys)	ALDH2	18.0/18.0	V1&V2&NEO	drug_response	0.1950	0.0002	0.0012	
INDEL	12:112241771	GA/G		frameshift_variant&splice_region_variant (p.Thr507fs)	ALDH2	18.0/18.0			0.0001			
SNV	12:112241795	T/C	rs147178045	intron_variant	ALDH2	18.0/17.9				0.0027		
SNV	12:112241839	C/T	rs533526749	intron_variant	ALDH2	16.7/16.7				0.0008		
SNV	12:112241840	G/A	rs781074160	intron_variant	ALDH2	16.7/16.7				0.0005		
SNV	12:112241860	G/A	rs879517880	intron_variant	ALDH2	16.9/16.9						
INDEL	12:112241867	TG/T	rs1160491288	intron_variant	ALDH2	17.2/17.2						
SNV	12:112241874	T/C	rs946163512	intron_variant	ALDH2	17.0/17.0				0.0004		
SNV	12:112241876	G/A	rs917600157	intron_variant	ALDH2	17.0/17.0				0.0004		

バリアントのQuality:
FILTER=PASSバリアントのQuality:
FILTER=not PASSAFR: African/African American
AMR: Admixed American
ASJ: Ashkenazi Jewish
EAS: East Asian
NFE: Non-Finnish European

ゲノムバリエント



Genome Variation

結果一覧→ブラウザ表示



Sequence



Variation



Methylome



Transcriptome



Proteome



Metabolome



Repository



GWAS

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Search by gene: ALDH2 (8.3KJPN)

GRCh37/hg19

4122 variants found

Filter by keyword

「Browser」をクリックしてブラウザを表示

Statistics

Browser

Plot

« ‹ … 29 30 31 … › »

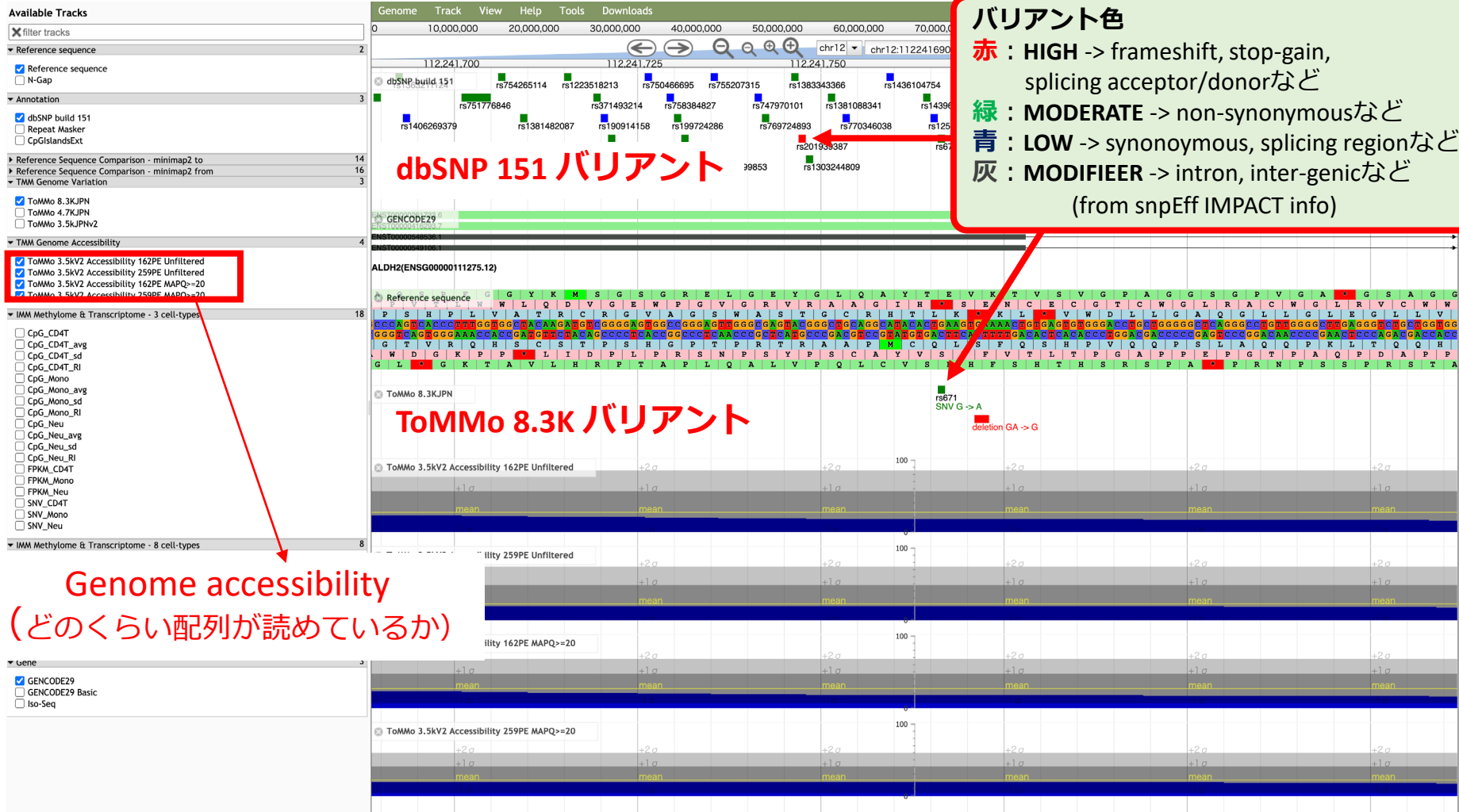
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ゲノムバリエント



Genome Variation

結果一覧→ブラウザ表示



ゲノムバリエント



Genome Variation

結果一覧→集団間の頻度比較

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



Search by gene: ALDH2 (8.3KJPN)

GRCh37/hg19

4122 variants found

「Plot」をクリックして集団間の頻度比較を表示

Browse

Plot

Filter by keyword

29 30 31

Type	Position ↑↓	Ref/Alt	rs#	Annotation ↑↓	Gene ↑↓	MeanDepth (162PE)	JPA ↑↓	ClinVar Annotation	ToMMo 8.3KJPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ
SNV	12:112241766	G/A	rs671 ↗	missense_variant (p.Glu504Lys ↗)	ALDH2 ↗	18.0/18.0	V1&V2&NEO	drug_response ↗	0.1950	0.0002	0.0012	
INDEL	12:112241771	GA/G		frameshift_variant&splice_region_variant (p.Thr507fs)	ALDH2 ↗	18.0/18.0			0.0001			
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SNV	12:112241874	T/C	rs946163512 ↗	intron_variant	ALDH2 ↗	17.0/17.0						
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ゲノムバリエント



Genome Variation

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結果一覧→集団間の頻度比較

[Back to table](#)Search by gene: ALDH2 (8.3KJPN) GRCh37/hg19

Panel1

ToMMo 8.3KJPN

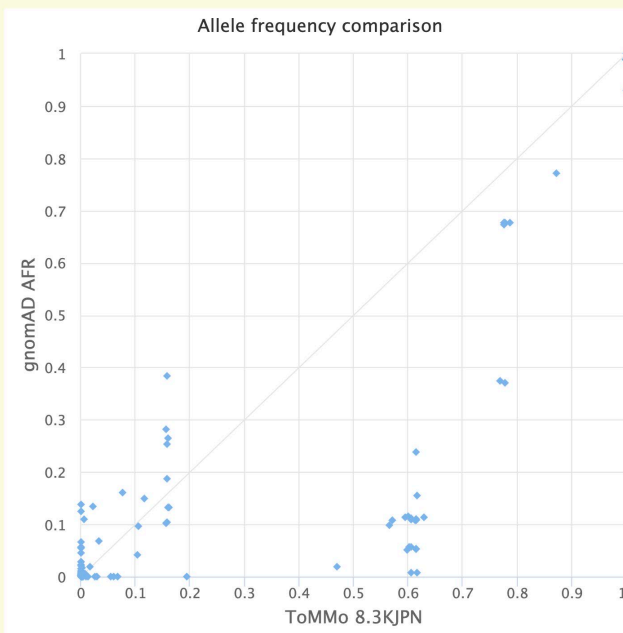
Panel2

gnomAD AFR

☐ Show ALL variants (NoCall is shown as MAF=0)

Annotation

- ☒ 3_prime_UTR_variant (500)
- ☒ 5_prime_UTR_premature_start_codon_gain_variant (1)
- ☒ 5_prime_UTR_variant (8)
- ☒ downstream_gene_variant (313)
- ☒ frameshift_variant (1)
- ☒ frameshift_variant&splice_region_variant (1)
- ☒ intergenic_region (427)
- ☒ intron_variant (844)
- ☒ missense_variant (29)
- ☒ splice_region_variant (1)
- ☒ splice_region_variant&intron_variant (2)
- ☒ splice_region_variant&intron_variant (1)
- ☒ synonymous_variant (18)
- ☒ upstream_gene_variant (433)



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 (8.3KJPN)

GRCh37/hg19

Statistics

4122 variants found

Filter by keyword

クリックして「タンパク質構造」を表示

Browser

Plot

Type	Position ↑↓	Ref/Alt	rs#	Annotation ↑↓	Gene ↓	MeanDepth (162PE)	JPA ↑↓	ClinVar Annotation	ToMMo 8.3KJPN	gnomAD AFR	gnomAD AMR	gnomAD ASJ
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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

Blast-Hit1 Blast-Hit2 Blast-Hit3 Blast-Hit4 Blast-Hit5 Blast-Hit6 Blast-Hit7 Blast-Hit8 Blast-Hit9 Blast-Hit10

Blast result

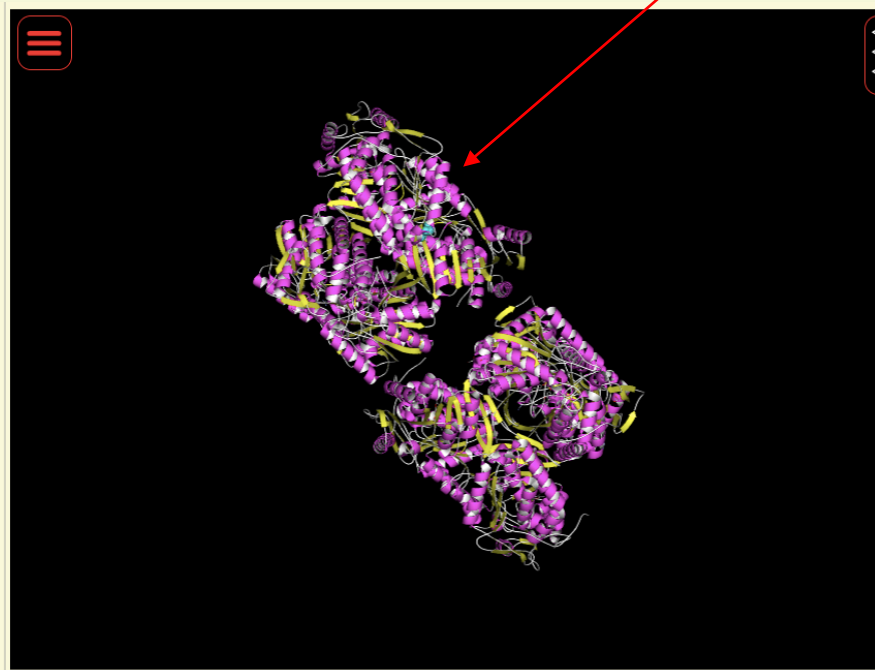
mRNA info	Homo sapiens aldehyde dehydrogenase 2 family (mitochondrial) (ALDH2), transcript variant 1, mRNA.
mRNA change	NM_000690.3:c.1510G>A_NP_000681.2:p.504E>K
Query	ALDH2 (GeneID: 217) NM_000690.3 NP_000681.2
Subject	5113
e-value	0.0000
Sequence Identity	100.00

BLAST結果選択

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)

マウスで
interactiveに
操作可能



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

二次構造

溶媒接触
表面積

複合体形成時の
溶媒接触表面積
変化

PDBファイルの
チェーン・残基