

バリエントデータの各項目について

1. バリエント (必須)
バリエントの情報は、遺伝子名とcoding DNAの変化 (c.) が基本情報になります。
2. 遺伝子; 参照配列 (必須)
mRNAのアクセッション番号 (NM_XXXあるいはENSTXXX) をご記載下さい。
不明な場合は検査実施施設にお問い合わせ下さい。
3. ゲノム上の物理位置
各参照配列 (hg19/hg38) に従った位置をご記載下さい。
不明な場合でもClinVarやdbSNPで検索可能です。
4. アミノ酸変化
アミノ酸の変化 (p.) が不明な場合やイントロン上のバリエントの場合は空欄で構いません。
データベース検索で明らかになった場合はご記載下さい。
5. ClinVarのID
「ClinVarのID検索方法 (資料1)」を参照してClinVarのIDを記載して下さい。
ClinVarにヒットしなかった場合は空欄で構いません。
6. dbSNPのID (rsXXX; rs番号)
アノテーション情報として分かっている場合にはご記載下さい。
「dbSNPの検索方法 (資料2)」を参照して内容を確認して下さい。
ClinVarで検索可能なバリエントの場合は「ClinVarのID検索方法 (資料1)」を参考にご記載下さい。
ClinVarにヒットしなかった場合、不明な場合は空欄で構いません。 rs番号が分かると検索が楽になります。
7. jMORP
「jMORPの検索方法 (資料3)」を参考にご記載下さい。
rs番号が不明な場合はゲノム上の物理位置でも検索できます。遺伝子名でも検索できますが、やや大変です。
不明な場合は空欄で構いません。
8. HGVD
rs番号が明らかな場合、「HGVDの検索方法 (資料4)」を参考にご記載下さい。不明な場合は空欄で構いません。
9. その他データベースの頻度情報
情報としてお持ちの場合にはご記載下さい。
「dbSNPの検索方法 (資料2)」からもgenomAD等の情報を検索する事が可能です。
必ずpopulationの情報を併記して下さい。

注意点

1. バリアントの表記
バリアントの表記はHGVS表記 (<http://varnomen.hgvs.org/>) に従って下さい。
困難な場合はvcfファイル上の表記をそのままご記載下さい。
MLPAの結果や、CNV等ゲノム上の物理位置が不明の場合はバリアントの表記も含めて個別に対応します。
エクソンや遺伝子名等で大まかにご記載下さい。
2. 参照配列の区別
検索を行う際には、参照配列の種類 (hg19あるいはhg38) にご注意下さい。
3. 頻度情報
頻度情報は、gnomAD-GenomesあるいはThe PAGE Studyの該当する民族のデータをご記載下さい。
どちらも無い場合には、ExAC等新しいものをご使用下さい。
4. バリアントの判定
バリアントの「判定の理由・根拠」欄には、公開されているデータベースでの判定 (あれば) を含めて下さい。
MLPA・CNVの結果等、データベースと正確に照らし合わせる事が困難な場合には、参考にしたデータを示して下さい。

統合データベースを用いた検索

ある程度慣れている方向けに、統合データベースを用いた検索方法を資料5と6に示します。
いずれも国内で整備されているデータベースですので、ご活用下さい。

- ・ 資料5 : MGeND (<https://togovar.biosciencedbc.jp/>)
- ・ 資料6 : TogoVar (<https://mgend.med.kyoto-u.ac.jp/>)

各データベースのリンク


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

jMORP (<https://jmorp.megabank.tohoku.ac.jp/201909/>)

dbSNP (<https://www.ncbi.nlm.nih.gov/snp/>)

HGVD (<http://www.hgvd.genome.med.kyoto-u.ac.jp/>)

ClinVarのID検索方法（資料1）



NCBI Resources How To Sign in to NCBI

ClinVar ClinVar Search **MSH2 c.340G>T と入力** Search

Advanced Help

Home About Access Help Submit Statistics FTP

```
ACTGATGGTATGGGGCCAAGAGATATATCT
CAGGTACGGCTGTCATCACTTAGACCTCAC
CAGGGCTGGGCATAAAAGTCAGGGCAG/
CCATGGTGCATCTGACTCCTGAGGAGA/
GCAGTTGGTATCAAGGTTACAAGACAC/
GGCACTGACTCTCTGCTATTGGTC/
```

ClinVar

遺伝子名 (MSH2) とバリアントの位置 (c.340G>T) を入力。
バリアントは正確に入力する。“340”や
“c.340”では検索できないので注意する。

Using ClinVar

[About ClinVar](#)[Data Dictionary](#)[Downloads/FTP site](#)[FAQ](#)[Contact Us](#)[RSS feed/What's new?](#)[Factsheet](#)[Submissions](#)[Variation Viewer](#)[Clinical Remapping - Between assemblies and RefSeqGenes](#)[RefSeqGene/LRG](#)[GTR®](#)[MedGen](#)[OMIM®](#)[Variation](#)

Submitter highlights

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

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

Want to learn more about who submits to ClinVar?

- [Read information about groups that submit to ClinVar](#)
- [See the list of submitters with the number of records each has submitted](#)

資料1

NCBI Resources How To

ClinVar Search

Create alert Advanced

Home About Access Help Submit Statistics FTP

Tabular

Download



There is one genomic location for MSH2:c.340G>T
See also [Variation Viewer](#)

Was this helpful?



| Location (GRCh38) | Variation | dbSNP | ClinVar |
|-------------------|----------------------|-----------------------------|------------------------------|
| chr2: 47,408,529 | NM_000251.3:c.340G>T | rs878853815 | VCV000237395 |

Search result

| Variation <i>Location</i> | Gene(s) | Protein change | Condition(s) | Clinical significance <i>(Last reviewed)</i> | Review status | Accession |
|---|----------------------|----------------|----------------|---|--|--------------|
| <input type="checkbox"/> 1. NM_000251.2(MSH2):c.340G>T (p.Glu114Ter) GRCh37: Chr2:47635668 GRCh38: Chr2:47408529 | MSH2 | E114*, E48* | Lynch syndrome | Pathogenic/Likely pathogenic <i>(Mar 30, 2018)</i> | criteria provided, multiple submitters, no conflicts | VCV000237395 |

該当するバリエントをクリック

ClinVar Genomic variation as it relates to human health [Search ClinVar](#)
Advanced search

[About](#) [Access](#) [Submit](#) [Stats](#) [FTP](#) [Help](#)

← [Switch to classic view](#) [Download](#) [Print](#) **ALPHA**

NM_000251.2(MSH2):c.340G>T (p.Glu114Ter) [Cite this record](#)

Interpretation: Pathogenic

Review status: ★☆☆☆ criteria provided, single submitter

Submissions: 1 (Most recent: Jun 10, 2016)

Last evaluated: Feb 14, 2016

Accession: VCV000237395.1

Variation ID: 237395 **ClinVarのID**

Description: single nucleotide variant

Variant details

Conditions

Gene(s)

NM_000251.2(MSH2):c.340G>T (p.Glu114Ter)

Allele ID: 238810

Variant type: single nucleotide variant

Variant length: 1bp

Cytogenetic location: 2p21

Genomic location: 2: 47408529 (GRCh38) [GRCh38 UCSC](#)
2: 47635668 (GRCh37) [GRCh37 UCSC](#) **ゲノム上の位置 (参照配列)**

HGVS:

| Nucleotide | Protein | Molecular consequence |
|---|---|-----------------------|
| NC_000002.12:g.47408529G>T | | |
| NC_000002.11:g.47635668G>T | | |
| NM_000251.2:c.340G>T | NP_000242.1:p.Glu114Ter | nonsense |

... more HGVS

Protein change: E114*

Functional consequence: -

Global minor allele frequency (GMAF): -

Allele frequency: -

Links: [dbSNP: rs878853815](#) **dbSNPのIDとリンク
リンク先を確認する**

FEEDBACK

**アクセション番号が正しい事を確認
アミノ酸変化はここに記載されている**

dbSNPの検索方法 (資料2)

例：MUTYH c.934-2A>G (rs77542170)

dbSNP

SNP rs77542170 Search

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

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

Access Data

Variation Services API

FTP Download

Tutorials on GitHub

April 8, 2019: dbSNP Entrez updates have been completed. The updates are listed here.

Notifications:

- As previously announced on (April 19, 2018), dbSNP Entrez currently only hot format will no longer be available. dbSNP Entrez eUtils will transition to a new 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

Display Settings: Summary

rs77542170 [*Homo sapiens*]

Variant type: SNV

Alleles: T>C [Show Flanks]

Chromosome: 1:45332088

Gene: MUTYH (Varview)

Functional Consequence: splice_acceptor_variant

Clinical significance: pathogenic-likely-pathogenic,conflicting-interpretations-of-pathogenicity,uncertain-significance

Validated: by frequency,by cluster

MAF: C=0.0007/86 (TOPMED)
C=0.0009/27 (GnomAD)
C=0.0010/124 (ExAC)
C=0.0011/285 (GnomAD_exomes)
C=0.0023/180 (PAGE_STUDY)
C=0.0030/15 (1000Genomes)

HGVs: NC_000001.11:g.45332088T>C, NC_000001.10:g.45797760T>C, NG_008189.1:g.13383A>G

PubMed LitVar

ID番号が必要

クリック

Reference SNP (rs) Report

ClinVarへのリンク

Switch to classic site

rs77542170

Current Build 153
Released July 9, 2019

Organism *Homo sapiens*
Position chr1:45332088 (GRCh38.p12)
Alleles T>C
Variation Type SNV Single Nucleotide Variation
Frequency C=0.00113 (285/251466, GnomAD_exome)
C=0.00068 (86/125568, TOPMED)
C=0.00102 (124/121300, ExAC) (+ 3 more)

Clinical Significance Reported in [ClinVar](#)
Gene : Consequence MUTYH : Splice Acceptor Variant
Publications 7 citations

バリアントの位置が正しい事を確認。
下の方にブラウザもあります。

Variant Details

Genomic Placements

Clinical Significance

Frequency

| Sequence name | Change |
|----------------------------|----------------------------|
| GRCh37.p13 chr 1 | NC_000001.10:g.45797760T>C |
| GRCh38.p12 chr 1 | NC_000001.11:g.45332088T>C |
| MUTYH RefSeqGene (LRG_220) | NG_008189.1:g.13383A>G |

各データベースでの頻度情報

Aliases
Submissions

MUTYH DNA glycosylase (minus strand)

| | Change | Amino acid[Codon] | SO Term |
|-----------------------------|-------------------|-------------------|-------------------------|
| MUTYH transcript variant 12 | NM_001350650.1:c. | N/A | Splice Acceptor Variant |
| MUTYH transcript variant 13 | NM_001350651.1:c. | N/A | Splice Acceptor Variant |

頻度情報

| | | | | | | |
|----------------------------------|----------------------------------|------------|--------|-----------|-----------|----------|
| gnomAD - Exomes | AMERICAN | Sub | 34300 | T=1.0000 | C=0.0000 | |
| gnomAD - Exomes | African | Sub | 16256 | T=1.0000 | C=0.0000 | |
| gnomAD - Exomes | Ashkenazi Jewish | Sub | 10080 | T=1.0000 | C=0.0000 | |
| gnomAD - Exomes | Other | Sub | 6138 | T=1.000 | C=0.000 | |
| gnomAD - Genomes | Global | Study-wide | 31354 | T=0.9991 | C=0.0009 | |
| gnomAD - Genomes | | | | 0.0000 | C=0.0000 | |
| gnomAD - Genomes | African | Sub | 8696 | T=1.000 | C=0.000 | |
| gnomAD - Genomes | East Asian | Sub | 1560 | T=0.983 | C=0.017 | |
| gnomAD - Genomes | Other | Sub | 1088 | T=1.000 | C=0.000 | |
| gnomAD - Genomes | American | Sub | 848 | T=1.00 | C=0.00 | |
| gnomAD - Genomes | Ashkenazi Jewish | Sub | 290 | T=1.00 | C=0.00 | |
| The PAGE Study | Global | Study-wide | 78690 | T=0.9977 | C=0.0023 | |
| The PAGE Study | | | | 0 | T=1.0000 | C=0.0000 |
| The PAGE Study | | | | 0 | T=1.0000 | C=0.0000 |
| The PAGE Study | Asian | Sub | 8318 | T=0.982 | C=0.018 | |
| The PAGE Study | PuertoRican | Sub | 7916 | T=1.000 | C=0.000 | |
| The PAGE Study | NativeHawaiian | Sub | 4532 | T=0.994 | C=0.006 | |
| The PAGE Study | Cuban | Sub | 4230 | T=1.000 | C=0.000 | |
| The PAGE Study | Dominican | Sub | 3826 | T=1.000 | C=0.000 | |
| The PAGE Study | CentralAmerican | Sub | 2450 | T=1.000 | C=0.000 | |
| The PAGE Study | SouthAmerican | Sub | 1982 | T=1.000 | C=0.000 | |
| The PAGE Study | NativeAmerican | Sub | 1260 | T=0.999 | C=0.001 | |
| The PAGE Study | SouthAsian | Sub | 856 | T=1.00 | C=0.00 | |
| TopMed | Global | Study-wide | 125568 | T=0.99932 | C=0.00068 | |

gnomAD(ExACの後継)

The PAGE Study

jMORPの検索方法（資料3）

様々なomicsデータを検索できる。“Genome Variation”を選択

The screenshot shows the jMORP website interface. At the top, there is a green header with the jMORP logo and the text "Welcome to Japanese Multi Omics Reference Panel." Below the header, there is a list of omics data categories represented by horizontal bars with icons and text:

- Phenome (To be provided)
- Metabolome
- Proteome
- Transcriptome (Iwate Medical Megabank Organization; iMethyl)
- Methylome (Iwate Medical Megabank Organization; iMethyl)
- Genome Variation** (highlighted with a red oval)
- Genome Sequence

On the right side of the page, there are two sections: "jMorp release 201909" and "jMorp Publication". The "jMorp release 201909" section contains two updates: "September 2nd, 2019 2019 Major Update" and "September 17th, 2019 Minor update". The "jMorp Publication" section contains three references.

At the bottom of the page, there is a footer with the following information: "jMorp release 201909 / LastUpdate: September 17th, 2019 / Conditions of Use", "Tohoku Medical Megabank Organization, Tohoku University", and "Contact: jmorp[at]omics.megabank.tohoku.ac.jp".

Sequence | Variation | Proteome

jMorp ~Japanese Multi Omics Reference Panel~

Genomic Variants

Search by gene name | Search by rs# | Search by region (GRCh37/hg19)

Gene symbol

Examples: ALDH2, NFE2L2, GATA1

Search

遺伝子名（かなり多くのバリエーションが検索される）、rs番号、ゲノム上の位置から検索できる。

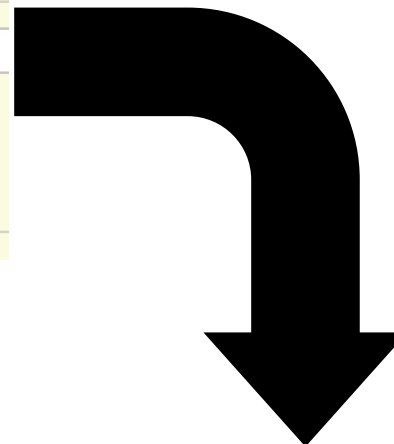
jMorp release 201909 / LastUpdate: September 17th, 2019 / Conditions of Tohoku Medical Megabank Organization, Tohoku University [↗](#)
Contact: jmorp [at] omics.megabank.tohoku.ac.jp

Genomic Variants

Search by gene name Search by rs# Search by region (GRCh37/hg19)

rsID:

Examples: rs671, rs6721961, rs1801133



Search by rs#: rs1799977 GRCh37/hg19

1 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 | 3:37053568 | A/G | rs1799977 | missense_variant (p.Ile219Val) | MLH1 | 29.0/29.0 | V1&V2 | Benign | 0.0489 | 0.0803 | 0.1611 | 0.2897 | 0.0205 | 0.3196 |

4.7KJPNにおける頻度や、gnomADでの頻度を見ることができる。

HGVDの検索方法（資料4）

Human Genetic Variation Database

[Home](#) [About](#) [Statistics](#) [Link](#) [Download](#) [Repository](#) [Contact](#) [How to Use](#) [Login](#)Welcome to [Human Genetic Variation Database](#)

Search database

Gene name/ID dbSNP rsID Pathogenic Variation

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

dbSNPのID (rs2303426) で検索

NGS Bioinformatics
Kyoto Course TOP GLOBAL
UNIVERSITY JAPANPage views: 3,449,223
Total downloads: 6,744

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](#)
- 02/15/2017 HGVD version 2.1 is available to browse and download.
- 02/25/2016 Our new paper "Human genetic variation database, a reference database of genetic variations in the Japanese population" has been published in *Journal of Human Genetics*. [How to cite](#)
- 07/02/2014 Bulk download of Cis-eQTL data is now available.
- 06/24/2014 Our paper "Large-Scale East-Asian eQTL Mapping Reveals Novel Candidate Genes for LD Mapping and the Genomic Landscape of Transcriptional Effects of Sequence Variants" has been published in *PLoS ONE*.
- 06/17/2014 Bulk download of genotype count data is now available.
- 11/12/2013 Web site has been created.

資料4

必ず「Org」が「All」となっているデータの頻度を記載する。

Exome

Allele Color Code: ■:A ■:G ■:C ■:T ■:Others

Variation Color Code: ■:nonsense, splice ■:missense ■:synonymous ■:indel :non-coding, intron, other

| Variant position | rsID | Ref/Alt | Frequency of alternative allele | Genotype count | | | Number of Samples Covered | Average sample read depth | Alleles | Gene | mRNA Accession# | Function | | | | Platform |
|------------------|-----------|---------|---------------------------------|----------------|---------|---------|---------------------------|---------------------------|--|------|-----------------|----------|-----|-------|---------------|----------|
| | | | | Ref/Ref | Ref/Alt | Alt/Alt | | | | | | Codon | | AA | | |
| | | | | | | | | | | | | Ref | Alt | Ref | Alt | |
| chr2:47630550 | rs2303426 | C/G | 0.7472 | 84 | 239 | 482 | 805 | 5.67±4.47 | 0.2528 ■ 0.7472 ■ | MSH2 | NM_000251 | intron | | All | | |
| chr2:47630550 | rs2303426 | C/G | 0.7394 | 27 | 93 | 162 | 282 | 7.35±5.66 | 0.2606 ■ 0.7394 ■ | MSH2 | NM_000251 | intron | | KU | HiSeq + SOLiD | |
| chr2:47630550 | rs2303426 | C/G | 0.7390 | 48 | 117 | 243 | 408 | 4.37±2.53 | 0.2610 ■ 0.7390 ■ | MSH2 | NM_000251 | intron | | YCU | HiSeq | |
| chr2:47630550 | rs2303426 | C/G | 0.7319 | 8 | 21 | 40 | 69 | 4.46±3.94 | 0.2681 ■ 0.7319 ■ | MSH2 | NM_000251 | intron | | NCCHD | HiSeq | |
| chr2:47630550 | rs2303426 | C/G | 0.8684 | 1 | 8 | 29 | 38 | 7.47±3.95 | 0.1316 ■ 0.8684 ■ | MSH2 | NM_000251 | intron | | TU | HiSeq | |
| chr2:47630550 | rs2303426 | C/G | 1.0000 | 0 | 0 | 8 | 8 | 14.88±7.79 | 0.0000 ■ 1.0000 ■ | MSH2 | NM_000251 | intron | | UT | HiSeq | |

MGeNDの検索方法（資料5）

The screenshot displays the MGeND website interface. At the top, there is a navigation menu with links for News, Statistics, Download, Source data, About us, Submission, and Help. The main heading is "Medical Genomics Japan Variants Database". Below this, a search bar contains the text "c.340G>T" and is circled in red. To the right of the search bar is a "Search" button. Below the search bar, an example is provided: "Example - Gene: BRAF, AA Change: E542K, Disease: Lynch syndrome". Below the search bar, there is a link for "Advanced Search" which is also circled in red. At the bottom, there are several category links: "Cancer", "Rare/Intractable diseases", "Infectious", "Dementia", and "Hearing loss".

遺伝子名、rs番号等、比較的フリーワードで検索できる。

Advanced Search

VCFやTSVフォーマットからもAnnotationを行う事が可能。

“All data”を選択することにより、よく使用されるデータベース上を全て検索できる。

MGeND Cancer ▾ Rare/Intractable diseases Infectious diseases ▾ Dementia Hearing loss ☰

MGeND data only **All data** SNV/INDEL (811) CNV (0) FUSION/SV (0) CLINICAL TRIAL (0)

Showing 1 to 2 of 2 entries (filtered from 811 total entries) Show entries

Free word

| Variant name | AA change | CDS | Japanese frequency | MGeND | | | ClinVar entry | ClinVar origin | ClinVar annotation | CIVIC evidence | DisGeNET entry | COSMIC occurrence |
|--|--------------|----------------------|--------------------|---------------|--------|------------------|---------------|----------------|--------------------|----------------|----------------|-------------------|
| | | | | Sample number | Origin | MGeND annotation | | | | | | |
| NC_000002.11:g.47635668G>T (rs878853815) | MSH2 p.E114* | NM_000251.2 c.340G>T | 〇〇〇 | 0 | | | 1 | | ★★★★ | 0 | 0 | |
| NC_000002.12:g.47408529G>T | MSH2 p.E114* | NM_000251.2 c.340G>T | 〇〇〇 | 0 | | | 1 | | ★★★★ | 0 | 0 | |

1

フリーワードで絞り込みが可能。

MGeND

Cancer ▾ Rare/Intractable diseases Infectious diseases ▾ Dementia Hearing loss ☰

MSH2 p.Glu114Ter (p.E114*) Detail

Summary Information Disease area statistics MGeND ClinVar CIVIC DisGeNET Annotation Genome browser

Summary


MGeND


Clinical significance

Variation sample number

GWAS sample number

Others

Clinical significance (ClinVar)  ★★★★★

Prediction(dbNSFP) 

Frequency(Japanese) [No Data.]

Entry(ClinVar) 1

Entry(CIVIC)

Entry(DisGeNET)

Disease name(ClinVar) Lynch syndrome

Information

| | |
|---------------------|---|
| Genome | hg19 |
| Position | chr2:47,635,668-47,635,668 |
| AA Change | p.E114* |
| HGVS | NC_000002.11:g.47635668G>T NM_000251.2:c.340G>T NP_000242.1:p.Glu114Ter |
| dbSNP | rs878853815 dbSNP |
| Gene symbol | MSH2 |
| Gene type | protein-coding |
| Gene description | mutS homolog 2 |
| Transcript ID | ENST00000233146.6 |
| Transcript position | chr2:47,630,108-47,710,367 |
| Transcript strand | + |
| MIM | 609309 OMIM |
| HGNC | HGNC:7325 HGNC |
| Ensembl | ENSG00000095002 Ensembl |

各種データベースへのリンクや、in silico predictionの結果、ブラウザを用いた周辺領域のバリエーションの確認等が行える。CIVICともリンクしており、somatic variantsについても検索可能。

TogoVarの検索方法（資料6）

rs番号での検索

TOGOVAR A comprehensive Japanese genetic variation database

Home Datasets Downloads Terms Contact About History Help Configuration

rs878853815

Disease: Breast-ovarian cancer, familial 2, MBO2, ovarian cancer, rs114202595 TogoVar: tgv421843 Position(GRCh37/hg19): 16:48258198 Region(GRCh37/hg19): 10:73270743-73376976

Results The number of available data is 10,000 out of 64,094,472.

| TogoVar ID | RefSNP ID | Position | Ref / Alt | Type | Gene | Alt frequency | Consequence | SIFT | PolyPhen | Clinical significance |
|-------------|--------------|----------|--------------|-----------|---------|---------------|---|------|----------|-----------------------|
| tgv67071957 | rs112750067 | 1: 10327 | T > C | SNV | | | Intergenic variant | | | |
| tgv67071958 | | 1: 10328 | A > | Deletion | | | Intergenic variant | | | |
| tgv67071959 | rs150969722 | 1: 10330 | C > | Deletion | | | Intergenic variant | | | |
| tgv67071961 | rs1351390918 | 1: 10334 | T > | Deletion | | | Intergenic variant | | | |
| tgv67072020 | rs376342519 | 1: 10617 | CGCC... 21bp | Deletion | | | Intergenic variant | | | |
| tgv67072021 | | 1: 10621 | GTTG... 27bp | Deletion | | | Intergenic variant | | | |
| tgv67072022 | | 1: 10622 | TT > | Deletion | | | Intergenic variant | | | |
| tgv67072033 | rs1266288166 | 1: 10816 | CA > | Insertion | | | Intergenic variant | | | |
| tgv67072075 | | 1: 12669 | AGAC... 7bp | Deletion | DDX11L1 | | Non coding transcript exon variant | | | |
| tgv67072091 | rs1290303072 | 1: 12868 | G > A | SNV | DDX11L1 | | Intron variant (+1) | | | |
| tgv67072092 | rs879918139 | 1: 12882 | C > G | SNV | DDX11L1 | | Intron variant (+1) | | | |
| tgv41 | rs62635286 | 1: 13116 | T > G | SNV | DDX11L1 | | Intron variant (+1) | | | |
| tgv42 | rs62028691 | 1: 13118 | A > G | SNV | DDX11L1 | | Intron variant (+1) | | | |
| tgv44 | rs531730856 | 1: 13273 | G > C | SNV | DDX11L1 | | Non coding transcript exon variant (+2) | | | |
| tgv47 | rs752859895 | 1: 13372 | G > C | SNV | DDX11L1 | | Splice region variant (+3) | | | |
| tgv53 | rs777038595 | 1: 13417 | GAGA > | Insertion | DDX11L1 | | Non coding transcript exon variant (+2) | | | |
| tgv54 | rs75175547 | 1: 13418 | G > A | SNV | DDX11L1 | | Non coding transcript exon variant (+2) | | | |
| tgv64 | rs574697788 | 1: 13494 | A > G | SNV | DDX11L1 | | Non coding transcript exon variant | | | |
| tgv66 | rs199896944 | 1: 13504 | G > A | SNV | DDX11L1 | | Non coding transcript exon variant | | | |
| tgv74 | rs775200581 | 1: 13539 | G > C | SNV | DDX11L1 | | Non coding transcript exon variant | | | |
| tgv79 | rs1263393206 | 1: 13657 | AG > | Deletion | DDX11L1 | | Non coding transcript exon variant (+1) | | | |
| tqv82 | rs796086906 | 1: 13868 | A > G | SNV | DDX11L1 | | Non coding transcript exon variant | | | |

Preview

Detailed variant report page

Genes Not found

External links refSNP: rs112750067

Alternative allele frequencies

| Dataset | Alt | Total | Frequency |
|---------|------|-------|-----------|
| JGA NGS | / | / | / |
| JGA SNP | / | / | / |
| 3.SJPN | 85 / | 5,438 | 0.0156 |
| HGVD | / | / | / |
| ExAC | / | / | / |

Consequence

Intergenic variant
A sequence variant located in the intergenic region, between genes

Clinical significance Not found

rs878853815

Disease: Breast-ovarian cancer, familial 2 Gene: ALDH2 RefSNP: rs11420255 TogoVar: tgv421843 Position: GRCh37:hg19: 16,482,58198 Region: GRCh37:hg19: 10,732,70743-73376976

Results The number of available data is 1 out of 1.

| TogoVar ID | RefSNP ID | Position | Ref / Alt | Type | Gene | All frequency | Consequence | SIFT | PolyPhen | Clinical significance |
|------------|-------------|------------|-----------|------|------|---------------|-------------|------|----------|-----------------------|
| tgv6780244 | rs878853815 | 2:47635668 | G>T | SNV | MSH2 | | Stop gained | | | Lynch syndrome |

TogoVar IDが割り振られている

Preview

Detailed variant report page

Genes

Symbol: MSH2
Alias: HNPCC, HNPCC1, DNA mismatch repair protein Msh2

External links

Variant report **tgv6780244** dbSNP rs878853815

ClinVarのデータやpopulation frequencyが簡便にまとめられている。

Other overlapping variant(s)

| TogoVar ID | Variant type | Ref / Alt | Consequence | SIFT | PolyPhen | Clinical Significance |
|------------|--------------|-----------|------------------------------|------|----------|-----------------------|
| tgv6780144 | deletion | GA | Splice acceptor variant (3*) | | | Lynch syndrome |

Frequency

No data

Clinical Significance

| Title | Clinical significance | Review status | Last evaluated | Condition(s) |
|--|-----------------------|---|----------------|----------------|
| NM_000251.2(MSH2):c.340G>T (p.Glu114Ter) | Pathogenic | ★★★★ criteria provided, single submitter | 2016-02-14 | Lynch syndrome |

Genomic context

Available Tracks

- DNA
- Gene
- Variant
- Frequency
- ExAC
- HGVD
- JGA-NGS
- JGA-SNP
- ToMMo

Genome Track View Help

0 20,000,000 40,000,000 60,000,000 80,000,000 100,000,000 120,000,000 140,000,000 160,000,000 180,000,000 200,000,000 220,000,000 240,000,000

47,635,625 47,635,650 47,635,675 47,635,700

tgv6780209 SNV G -> T
tgv6780207 SNV C -> T
tgv6780213 SNV A -> C
tgv67076289 SNV A -> C
tgv6780224 SNV T -> C
tgv6780228 SNV C -> A
tgv6780229 SNV C -> G
tgv6780236 SNV A -> C
tgv6780250 deletion ATTGGAT -> A
tgv6780251 SNV T -> C
tgv6780252 SNV G -> T,SNV G -> A
tgv6780263 SNV G -> T