

# 解析例 2

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CASと各種データベースを使用して  
疾患関連性を調べたLossの例

# Webinarの構成

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- インTRODクシヨン
  - マイクロアレイ染色体検査 (CMA) 検査 超入門
  - BEDファイル形式データの取扱い
  - マイクロアレイ染色体検査の結果解釈の補助ソフトウェアツール (CAS) の使用方法
  - マイクロアレイ (CMA) 検査の結果解釈に必要なデータベースの使用法
- 実践編
  - 解析例 1 : CASを使用して疾患関連性を調べたLossの例
  - 解析例 2 : CASと各種データベースを使用して疾患関連性を調べたLossの例
  - 解析例 3 : CASと各種データベースを使用して疾患関連性を調べたGainの例
  - 解析例 4 : 疾患関連性領域近傍にあるBenignのLossの例
- Advanced編
  - データベース「UCSC」使用法
  - 解析例 5 : UCSCを用いて、Uncertain Significance と推定される例
  - インプリンティング疾患の解釈について

# 免責事項

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- 実際の診断に際しては、個々の臨床所見と検査報告書並びにデータベース・ソフトウェアの検索結果を相互的に検討し判断を行ってください。
- 本セミナーで紹介するデータベース・ソフトウェアの使用法の正確性、妥当性について、演者、演者所属組織、本コンソーシアム関係者は一切の責任を負いません。
- 本セミナーで紹介する各例はあくまでもデータベースの使用例を提示するために疑似的に作成された例であり、実際の臨床情報並びに検査結果に基づいた例ではありません。
- 各データベース・ソフトウェアの使用について、そのデータの正確性、臨床的妥当性は保証されておらず、各データベース・ソフトウェアの作成者・管理者・研究班は提示される結果について一切の法的責任を負っておりません。

## 解析例 #2

chr20: 61830372-62298607 (ロス)

### ゲイン/ロス領域表 Gain/Loss Intervals Table

染色体 Chr	最小始点-終点(bp) 最大始点-終点(bp) Min Start-Stop(bp) Max Start-Stop(bp)	最小サイズ(kb) 最大サイズ(kb) Min Size(kb) Max Size(kb)	最小サイトバンド 最大サイトバンド Min Cytoband Max Cytoband	ゲイン/ロス Gain/Loss
chr20	61830372-62298607 61778582-62342175	468.235 563.595	q13.33 q13.33	-1.000

解析フロー：下記サイトを使用して検索します。

1. CASによる解析
2. DECIPHER
3. ClinGen Dosage Sensitivity

## 解析に有用なURL

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マイクロアレイ染色体検査の結果解釈の補助ソフトウェアツール (CAS)

<https://cmg.med.keio.ac.jp/arrayclassified/>

DECIPHER

<https://www.deciphergenomics.org/>

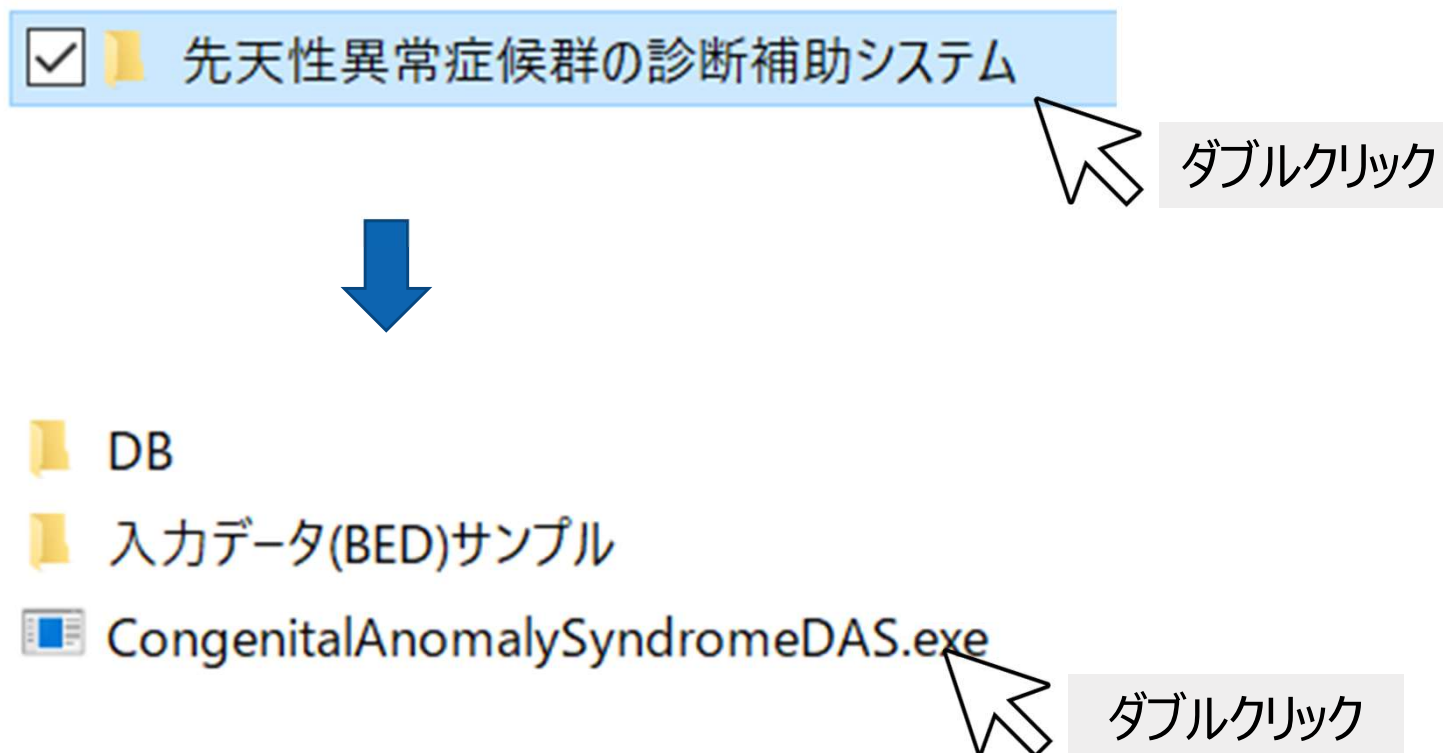
ClinGen Dosage Sensitivity

<https://www.clinicalgenome.org/>

# CASによる解析

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- 「先天性異常症候群の診断補助システム」フォルダの中にある CongenitalAnomalySyndromeDAS.exe を起動します。

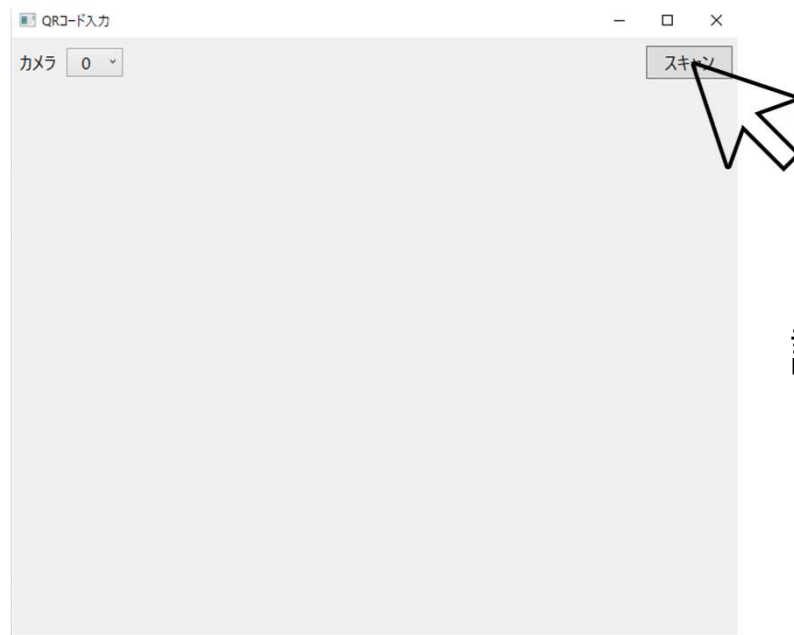


解析例2



# CASによる解析（読み込み①）

- 報告書に印字される二次元バーコードをスキャンします。



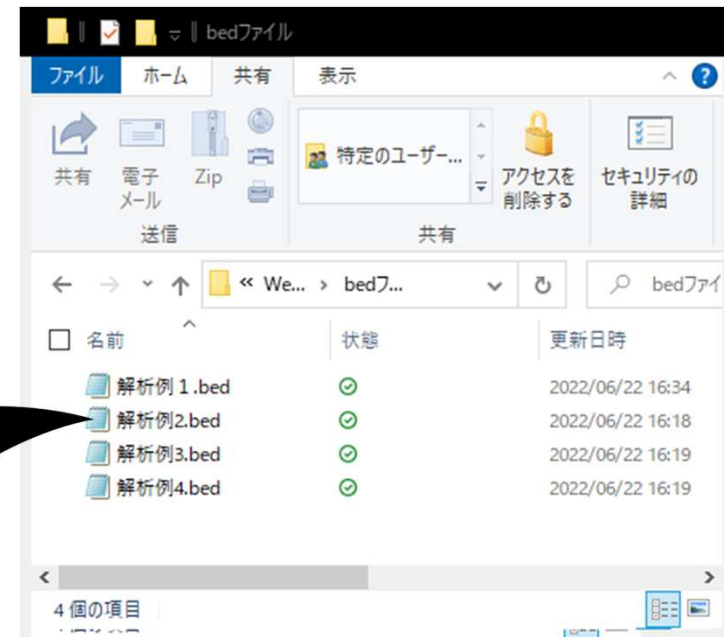
- パソコンのカメラから報告書の二次元バーコードを読み込みます。

解析例2

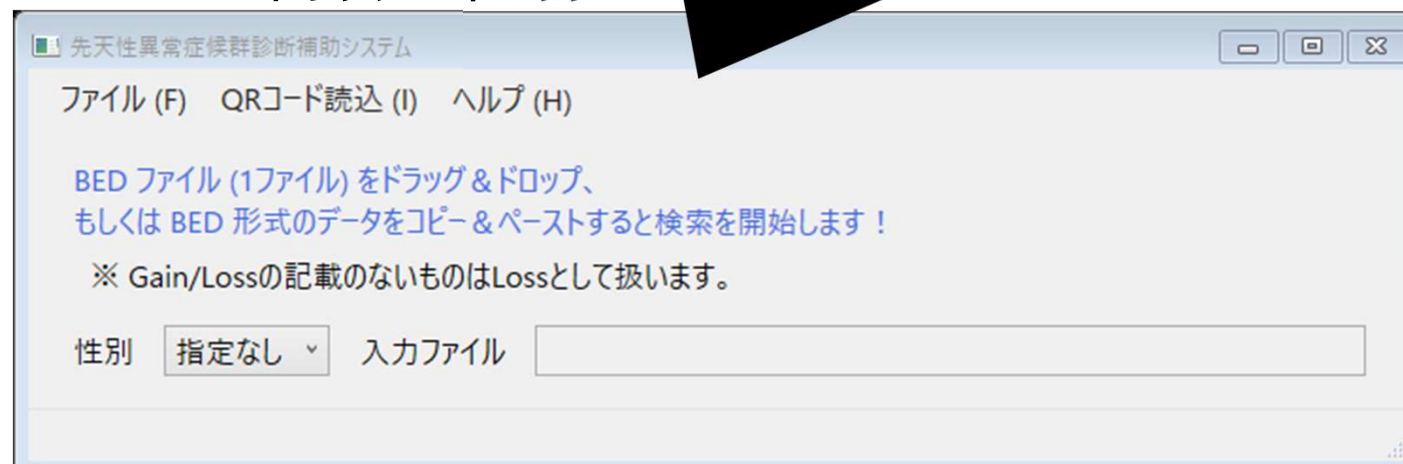


# CASによる解析（読み込み②）

- BEDファイルをウィンドウ内にドラッグ＆ドロップし、検索を開始します。



ドラッグ＆ドロップ



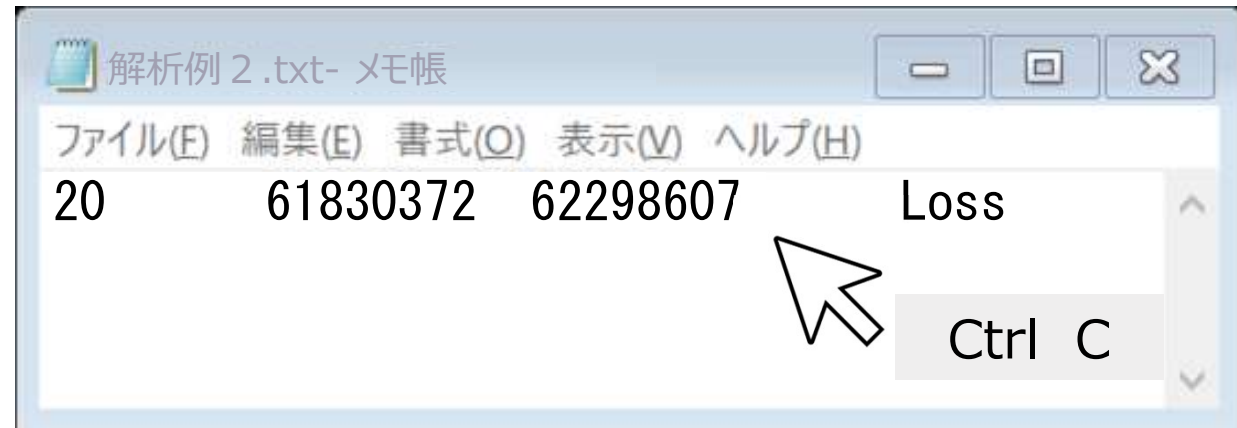
解析例2





# CASによる解析（読み込み③）

- BED形式のデータをコピー & ペーストすることでも検索ができます。



解析例2



# CASの解析結果①

症候群は該当しませんでした。

ロスの領域と重複するpLIスコアが0.9以上の遺伝子として、*YTHDF1*、*KCNQ2*、*EEF1A2*、*GMEB2*が表示されました。（そのうち領域が100%オーバーラップするものは3遺伝子でした。）

No.	染色体	開始	終了	Decipher browser	検索結果	症候群	染色体	開始	終了	遺伝子名	OMIM	染色体	開始	終了	重複範囲	スコア (pLI)
1	20	61,830,372	62,298,607	<a href="#">ブラウザへのリンク</a>	Match					YTHDF1	<a href="#">OMIM: 616529 (YTH N6-METHYLADENOSINE RNA-BINDING PROTEIN 1; YTHDF1)</a>	20	61,826,781	61,847,586	82.7 %	0.994
2	20	61,830,372	62,298,607	<a href="#">ブラウザへのリンク</a>	Match					KCNQ2	<a href="#">OMIM: 602235 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2; KCNQ2)</a>	20	62,037,542	62,103,993	100.0 %	1.000
3	20	61,830,372	62,298,607	<a href="#">ブラウザへのリンク</a>	Match					EEF1A2	<a href="#">OMIM: 602959 (EUKARYOTIC TRANSLATION ELONGATION FACTOR 1, ALPHA-2; EEF1A2)</a>	20	62,119,366	62,130,505	100.0 %	0.996
4	20	61,830,372	62,298,607	<a href="#">ブラウザへのリンク</a>	Match					GMEB2	<a href="#">OMIM: 607451 (GLUCOCORTICOID MODULATORY ELEMENT-BINDING PROTEIN 2; GMEB2)</a>	20	62,218,955	62,258,394	100.0 %	0.995

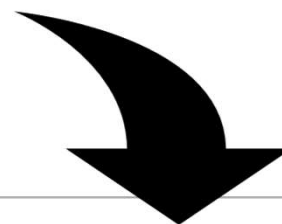
解析例2



## CASの解析結果②

それぞれの検出された遺伝子には、OMIMページのリンクが表示されますので、参照することができます。

遺伝子名	OMIM	染色体	開始	終了	重複範囲	スコア (pLI)
YTHDF1	<a href="#">OMIM: 616529 (YTH N6-METHYLADENOSINE RNA-BINDING PROTEIN 1; YTHDF1)</a>	20	61,826,781	61,847,586	82.7 %	0.994
KCNQ2	<a href="#">OMIM: 602235 (POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2; KCNQ2)</a>	20	62,037,542	62,103,993	100.0 %	1.000
EEF1A2	<a href="#">OMIM: 602959 (EUKARYOTIC TRANSLATION ELONGATION FACTOR 1, ALPHA-2; EEF1A2)</a>	20	62,119,366	62,130,505	100.0 %	0.996
GMEB2	<a href="#">OMIM: 607451 (GLUCOCORTICOID MODULATORY ELEMENT-BINDING PROTEIN 2; GMEB2)</a>	20	62,218,955			



\* 602235

**POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2; KCNQ2**

*Alternative titles; symbols*  
 POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY Q, MEMBER 2

*HGNC Approved Gene Symbol: **KCNQ2***

*Cytogenetic location: 20q13.33 Genomic coordinates (GRCh38): 20:63,400,207-63,472,654 (from NCBI)*

**Gene-Phenotype Relationships**

Location	Phenotype <span style="background-color: #FFD700;">Clinical Synopses</span>	Phenotype MIM number	Inheritance	Phenotype mapping key
20q13.33	Developmental and epileptic encephalopathy 7	613720	AD	3
	Myokymia	121200	AD	3
	Seizures, benign neonatal, 1	121200	AD	3



# DECIPHER

https://www.deciphergenomics.org/

DECIPHER v11.11: Mapping the clinical genome

https://www.deciphergenomics.org

DECIPHER GRCh38 About Browse DDD (UK) Search DECIPHER Help Join Log in

## Mapping the clinical genome

### Explore DECIPHER

It's free and you don't need to log in

DECIPHER is used by the clinical community to share and compare phenotypic and genotypic data. The DECIPHER database contains data from 42,711 patients who have given consent for broad data-sharing; DECIPHER also supports more limited sharing via consortia. [Have a look at the numbers.](#)

Anyone can browse publicly-available patient data on DECIPHER and request to be put in contact with the responsible clinician. Why? [Because sharing benefits everyone.](#)

[Explore DECIPHER's genome browser](#)

Delve into the Human Phenotype Ontology

### Join DECIPHER

Be part of the sharing community

Projects affiliated to DECIPHER can deposit and share patients, variants, and phenotypes to invite collaboration and facilitate diagnosis. Once deposited, you can use DECIPHER to identify and prioritise potential matches, and you can request notifications as soon as new matches arrive.

As well as influencing individual patient outcomes, use of DECIPHER has contributed to over [2600 published articles since 2004](#). It's still free, and you are in control of what data to make public.

[Join now](#)

Find out more

### Already a member?

Log in to access your patient data

**Email address**

**Password**

[Log in](#)

Reset your password

Feedback



# DECIPHERの検索

DECIPHERのデフォルト座標はGRCh38です。本検査結果はGRCh37の座標で示されているので、明示的に示す必要があります。

例) grch37:20:61830372-62298607

DECIPHER v11.11: Mapping the c x +

https://www.deciphergenomics.org

grch37:20:61830372-62298607

e.g. EP300, Hypertrichosis, splice\_donor\_variant, more... Clear filters

クリック

Close Clear filters Reset filters Search

Search examples

You can use any of the following to search for patients and variants deposited into DECIPHER.

- [Hypertrichosis](#) - by phenotype
- [hp:0001831](#) - by HPO Identifier. Prefix the HPO ID you wish to search with **hp**:
- [6:157099063-157531913](#) - by position in GRCh38
- [grch37:6:157099063-157531913](#) - by position in GRCh37
- [17p11.2](#) - by band (band will be converted to position)
- [EP300](#) - by gene
- [Benign](#) - by **pathogenicity**
- [Biparental](#) - by **inheritance**
- [splice\\_donor\\_variant](#) - by **consequence**
- [255882](#) - by DECIPHER patient ID
- [dendritic spines](#) - plain text search in syndrome names, descriptions, etc

EXACT VARIANT SEARCHES

Searches for small DNA sequence and protein variants will take you to a dedicated page for that change (whether or not a DECIPHER variant exists). These searches cannot be combined with other terms.

- [X:77652333 CTCT>C](#) - DNA sequence variant by GRCh38 location (1-based)
- [22:41177728:AC:](#) - DNA sequence variant by SPDI (0-based; use three colons)
- [chrX:g.7257548\\_7257549insA](#) - DNA sequence variant by HGVSg

GVSc and HGNC gene symbol  
GVSc and RefSeq transcript  
HGNC gene symbol  
(graphs) and RefSeq transcript

Note that Ensembl stable IDs (ENSG, ENST, ENSP) are also accepted in place of other identifiers.

You can combine multiple terms (except variant searches), by separating them with a comma and a space, for example:

- [17p11.2, Dyscalculia](#) - Combine band and phenotype
- [Arachnodactyly, High palate](#) - Search for patients with more than one phenotype

# DECIPHERの検索結果

CNV syndrome variants と Genesの結果を確認します。

DECIPHER CNV syndrome variantsの結果は0ヒットでしたので、「確立されたCNVシンドロームなし」と考えられます。

DECIPHER v11.11: Mapping the c x +

https://www.deciphergenomics.org/search/patients/results?q=grch37:20:61830372-62298607 GRCh38 20:63199020-63667254 (95% match) (Refine Search)

Patient variants 177 CNV syndrome variants 0 DDD research variants 5 Genes 22

Results Browser

Variants: 1 to 10 of 177 Show: All variant types Filter...

DECIPHER Patient	Sex	Location	Size	Inheritance / Genotype	Pathogenicity / Contribution	Phenotype(s)	Contact
635	46XY	20 <sup>56777975</sup> <sub>64182879</sub> GRCh38 Duplication	7.40 Mb	Unknown Heterozygous		Hypotonia, Intellectual disability, Microcephaly	✉
771	46XX	20 <sup>60872006</sup> <sub>64291218</sub> GRCh38	3.42 Mb	De novo Heterozygous		Abnormality of the upper limb, Coarctation of aorta, Hypoplastic philtrum, Hypotonia, Intellectual disability, Protruding tongue, Short phalanx of	✉



# DECIPHER Genes の結果

ClinGenの項目の矢印をクリックし、ClinGenのcuration activitiesに関する数値で並び替えます。

The screenshot shows the DECIPHER Genes page with a table of genes. The 'ClinGen' column has a dropdown arrow highlighted with a red box. A popup window titled 'ClinGen' is open, displaying information about curation activities and a dosage sensitivity rating table.

Name / Description	Location	pLI	LOEUF	sHet	%HI	GenCC	OMIM / Morbid	G2P	ClinGen	Links
ARFGAP1 ADP ribosylation factor GTPase activating protein 1	20 63272785 63289790	0.00	0.63	0.113	66.43	-	OMIM	-	-	View
BIRC7 baculoviral IAP repeat co										View
CHRNA4 cholinergic receptor nico										View

**ClinGen**

ClinGen has a number of curation activities related to defining the clinical relevance of genes and variants for use in precision medicine and research.

**Gene/disease validity**

The [ClinGen Gene Curation working group](#) has developed a framework to standardize the approach to determine the clinical validity for a gene-disease pair.

Gene-disease classifications are: definitive, strong, moderate, limited, no reported evidence, refuted and disputed.

Further information is available [here](#).

**Dosage Sensitivity**

The [ClinGen Dosage Sensitivity working group](#) collects evidence supporting/refuting the haploinsufficiency and triplosensitivity of genes and genomic regions.

Further information is available [here](#).

Dosage sensitivity rating	Possible clinical interpretation
3	Sufficient evidence for dosage pathogenicity
2	Some evidence for dosage pathogenicity
1	Little evidence for dosage pathogenicity
0	No evidence for dosage pathogenicity
40	Evidence suggests the gene is not dosage sensitive (haploinsufficiency or triplosensitivity is unlikely)
30	Gene associated with autosomal recessive condition

OK

解析例2



# DECIPHER Genes の結果 (ClinGenの項目で並び替え後)

ハプロ不全により発症するものとしてClinGenのcurationがなされている (ClinGen Haploinsufficiency: 3) *KCNQ2*が検索されました。

その他にも、pLIの数値が高く、ハプロ不全が想定される遺伝子として*EEF1A2*も検索されました。%HIも併せて画面上で確認することができます。

Patient variants 177    CNV syndrome variants 0    DDD research variants 5    Genes 22

Genes: 1 to 10 of 22    Show: All genes    Filter...

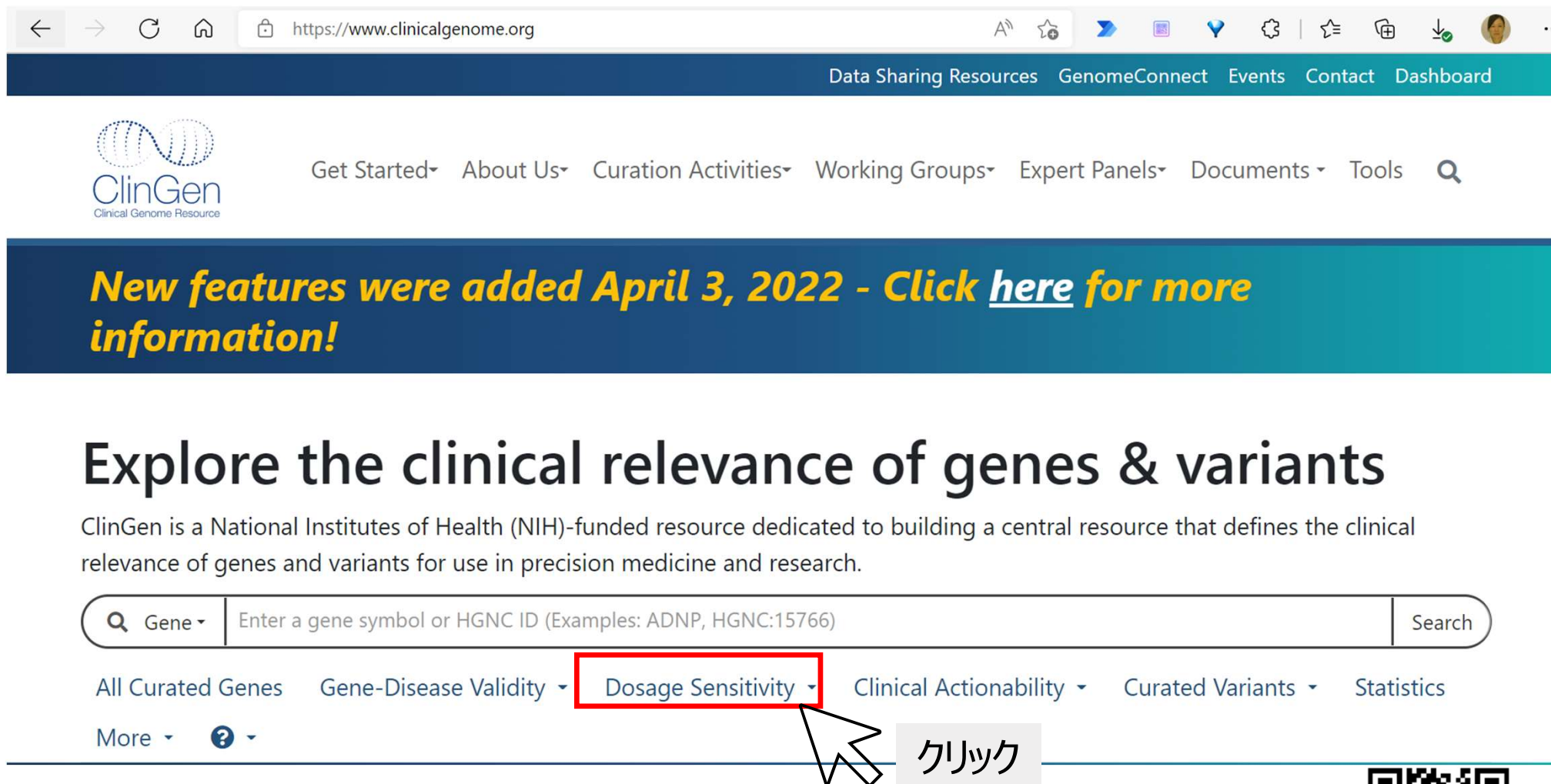
Name / Description	Location	pLI	LOEUF	sHet	%HI	GenCC	OMIM / Morbid	G2P	ClinGen	Links
<i>CHRNA4</i> cholinergic receptor nicotinic alpha 4 subunit	20 63343223 63378401	0.00	0.82	0.008	40.98	Definitive: Strong: Supportive:	2 1 1 OMIM Morbid (2)	Definitive: Monoallelic	Definitive: AD	View
<i>EEF1A2</i> eukaryotic translation elongation factor 1 alpha 2	20 63488013 63499239	1.00	0.19	0.174	36.28	Definitive: Moderate: Supportive:	1 1 2 OMIM Morbid (2)	Strong: Monoallelic	Definitive: AD	View
<i>KCNQ2</i> potassium voltage-gated channel subfamily Q member 2	20 63400208 63472677	1.00	0.16	0.138	39.78	Definitive: Supportive:	3 6 OMIM Morbid (2)	Definitive: Monoallelic	Haploinsufficiency: 3 Triplosensitivity: 0	View
<i>YTHDF1</i> YTH N6-methyladenosine RNA binding protein 1	20 63195429 63216139	0.99	0.24	0.216	62.30	-	OMIM	-	-	View





# ClinGen Dosage Sensitivityを用いた検索

ClinGenのDosage Sensitivityでも同様に検索をします。



The screenshot shows the ClinGen website interface. At the top, there is a navigation bar with links for "Data Sharing Resources", "GenomeConnect", "Events", "Contact", and "Dashboard". Below this is the ClinGen logo and a main navigation menu with links for "Get Started", "About Us", "Curation Activities", "Working Groups", "Expert Panels", "Documents", and "Tools". A prominent yellow banner reads: "New features were added April 3, 2022 - Click [here](#) for more information!". The main heading is "Explore the clinical relevance of genes & variants". Below this is a paragraph describing ClinGen as a NIH-funded resource. A search bar is present with the placeholder text "Enter a gene symbol or HGNC ID (Examples: ADNP, HGNC:15766)". Below the search bar is a navigation menu with several options: "All Curated Genes", "Gene-Disease Validity", "Dosage Sensitivity", "Clinical Actionability", "Curated Variants", and "Statistics". The "Dosage Sensitivity" option is highlighted with a red box, and a mouse cursor is pointing at it. A grey box with the Japanese word "クリック" (click) is positioned next to the cursor. A QR code is located in the bottom right corner of the screenshot.



# ClinGen Dosage Sensitivityの検索画面

「chr20:61830372-62298607」と入力して、検索をします。

Data Sharing Resources GenomeConnect Events Contact Login

ClinGen Clinical Genome Resource

Get Started About Us Curation Activities Working Groups Expert Panels Documents & Announcements Tools

Gene Enter a gene symbol or HGNC ID (Examples: ADNP, HGNC:15766) Search

All Curated Genes Gene-Disease Validity Dosage Sensitivity Clinical Actionability Curated Variants Statistics Downloads More

**D** Dosage Sensitivity Genes: On Regions: On

3726 Total Curations 1493 Total Genes 507 Total Regions

Advanced Filters: None Click on below to view hidden columns

Search in table Search in table Enter cytoband or genomic coordinates Go!

Showing 1 to 25 of 2000 rows 25 rows per page

GRCh37 GRCh38

① GRCh37を選択

② クリック

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Last Eval.
16p13.12 population region (gnomAD-SV_v2.1_DEL_16_152599)	16:14782199-14805000	40 (Dosage Sensitivity Unlikely)	0 (No Evidence)			-	-	-	08/09/2021



# ClinGen Dosage Sensitivityの結果

下記のように、23遺伝子、0領域が検出されます。

GRCh37 Search Results  
Location: chr20:61830372-62298607

Genes: On Regions: On

23 Total Genes 0 Total Regions

Advanced Filters: None

Search in table GRCh37 Enter cytoband or genomic coordinates Go!

Showing 1 to 23 of 23 rows 25 rows per page

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
YTHDF1	20 61826781 61847538	Not Yet Evaluated	Not Yet Evaluated	✓		62.31	0.99	0.24	Awaiting Review
BIRC7	20 61867235 61871859	Not Yet Evaluated	Not Yet Evaluated	✓		87.65	0	1.34	Awaiting Review
MIR3196	20 61870131 61870194	Not Yet Evaluated	Not Yet Evaluated			-	-	-	Awaiting Review
NKAIN4	20 61872136 61885892	Not Yet Evaluated	Not Yet Evaluated	✓		79.72	0.01	0.99	Awaiting Review
FLJ16779	20 61885330 61892967	-1 (Pseudogene)	-1 (Pseudogene)			-	-	-	Not Reviewable
AREGAP1	20 61904137	Not Yet Evaluated	Not Yet Evaluated	✓		66.43	0	0.6	Awaiting Review

列2



# ClinGen Dosage Sensitivity Genesの結果

遺伝子の情報をOn に、領域の表示をOffに設定します。

画面右上の 🔍 をクリックし Column Searchを表示させます。

The screenshot shows the ClinGen GRCh37 Search Results interface. At the top, there are toggle buttons for 'Genes: On' and 'Regions: Off', both highlighted with a red box. A mouse cursor points to the 'Genes: On' button. To the right, a text box says 'クリックして、表示・非表示を切り替え' (Click to toggle display/non-display). Below this, another red box highlights a magnifying glass icon in the top right toolbar, with a mouse cursor pointing to it. The main content area shows a table with columns for Gene/Region, GRCh37, HI Score, TS Score, OMIM, Morbid, %HI, pLI, LOEUF, and Report. Three rows are visible: YTHDF1, BIRC7, and MIR3196. The table is titled 'GRCh37 Search Results' with location 'chr20:61830372-62298607'. There are also search filters and a search bar at the top.

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
YTHDF1	20 61826781 61847538	Not Yet Evaluated	Not Yet Evaluated	✓		62.31	0.99	0.24	Awaiting Review
BIRC7	20 61867235 61871859	Not Yet Evaluated	Not Yet Evaluated	✓		87.65	0	1.34	Awaiting Review
MIR3196	20 61870131 61870194	Not Yet Evaluated	Not Yet Evaluated			-	-	-	Awaiting Review



# ClinGen Dosage Sensitivity Genesの結果 (HI)

Haploinsufficiency Scoreの項目をクリックし、HI Score 3 (Sufficient Evidence) を選択すると、*KCNQ2*が検出されます。

GRCh37 Search Results  
Location: chr20:61830372-62298607  
Genes: On Regions: Off  
23 Total Genes, 0 Total Regions  
Advanced Filters: None  
Click on below to view hidden columns  
Search in table  
GRCh37 Enter cytoband or genomic coordinates Go!  
Showing 1 to 23 of 23 rows 25 rows per page

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
YTHDF1	20 61826761 61847538	-1 (Pseudogene) 3 (Sufficient Evidence) Not Yet Evaluated	Not Yet Evaluated	✓		62.31	0.99	0.24	Awaiting Review
BIRC7	20 61867235 61871859	Not Yet Evaluated	Not Yet Evaluated	✓		87.65	0	1.34	Awaiting Review



GRCh37 Search Results  
Location: chr20:62031567-62104030  
Genes: On Regions: Off  
23 Total Genes, 0 Total Regions  
Advanced Filters: None  
Click on below to view hidden columns  
Search in table  
GRCh37 Enter cytoband or genomic coordinates Go!  
Showing 1 to 1 of 1 rows 25 rows per page

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
KCNQ2	20 62031567 62104030	3 (Sufficient Evidence)	0 (No Evidence)	✓	✓	39.78	1	0.16	Complete

解析例2





# ClinGen Dosage Sensitivity Genesの結果 (Report)

Completeをクリックすると、Evidenceが表示されます。

Gene/Region	GRCh37	HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
KCNQ2	20 62031567 62104030	3 (Sufficient Evidence)	0 (No Evidence)	✓	✓	39.78	1	0.16	<a href="#">Complete</a>

Showing 1 to 1 of 1 rows

### Haploinsufficiency (HI) Score Details

**HI Score:** 3

**HI Evidence Strength:** Sufficient Evidence for Haploinsufficiency (Disclaimer)

**HI Disease:** seizures, benign familial neonatal, 1 [Monarch](#)

**HI Evidence:** [PUBMED: 17675531](#)  
Heron et al. (2007): Describes 3 intragenic deletions (all removing multiple continuous exons) of KCNQ2 detected amongst families affected with benign familial neonatal seizures (BFNS). An intragenic duplication of exons 3-12 was also described. These mutations were also detected in all affected relatives of each proband.

[PUBMED: 14534157](#)  
Singh et al. (2003): Describes a deletion extending from intron 8 of KCNQ2 through 22.1 kb past the KCNQ2 stop codon, deleting the last nine exons of KCNQ2. The authors propose that the "likely disease-causing mechanism is haploinsufficiency of the KCNQ2 protein, because mRNA produced from the deleted KCNQ2 allele lacks the poly A tail and is potentially degraded rapidly." Also describe 4 additional nonsense mutations detected in individuals with BFNS. The authors state that "these mutations cause a variable loss of function, and selective effects on the biophysical properties of KCNQ2/KCNQ3 heteromultimeric channels."

Of note, this paper also describes "the first dominant negative mutation in KCNQ2 that has a phenotype of neonatal seizures without permanent clinical CNS impairment" (867insGGGCC).

**HI Evidence Comments:** From Singh et al. (2003) (PMID:14534157): "Expression of a few of the mutations identified to date suggests that a partial loss of function in potassium current is sufficient to produce an epilepsy phenotype, and dominant negative mutations in either KCNQ2 or KCNQ3 may lead to a more severe phenotype (Jentsch, 2000, PMID:11252765)"

「家族性の  
良性てんかんの  
原因遺伝子」と  
記載がありますが  
個々の臨床所見  
と合わせて  
判断してください。

列2



# ClinGen Dosage Sensitivity Genesの結果 (OMIM)

OMIMをクリックすると、その登録内容を確認することができます。

 **KCNQ2**

Gene Facts

3 Haplo Score  
0 Triplo Score

Gene Facts [External Data Attribution](#)

HGNC Symbol KCNQ2 (HGNC:6296) [HGNC](#) [Entrez](#) [Ensembl](#) [OMIM](#) [UCSC](#) [Uniprot](#) [GeneReviews](#) [LOVD LSDB](#) [ClinVar](#)

HGNC Name potassium voltage-gated channel subfamily Q member 2

Gene type protein-coding gene

Locus type gene with protein product

Previous symbols EBN, EBN1

Alias symbols Kv7.2, ENB1, BFNC, KCNA11, HNSPC

%HI 39.78 [\(Read more about the DECIPHER Ha\)](#)

pLI 1 [\(Read more about gnomAD pLI score\)](#)

\* 602235

POTASSIUM CHANNEL, VOLTAGE-GATED, KQT-LIKE SUBFAMILY, MEMBER 2; KCNQ2

*Alternative titles; symbols*

POTASSIUM CHANNEL, VOLTAGE-GATED, SUBFAMILY Q, MEMBER 2

*HGNC Approved Gene Symbol: **KCNQ2***

*Cytogenetic location: 20q13.33 Genomic coordinates (GRCh38): 20:63,400,207-63,472,654 (from NCBI)*

**Gene-Phenotype Relationships**

個々の臨床所見と合わせて判断してください。

