

# データベース「UCSC」使用方法

埼玉県立小児医療センター 遺伝科

大場大樹

# Webinarの構成

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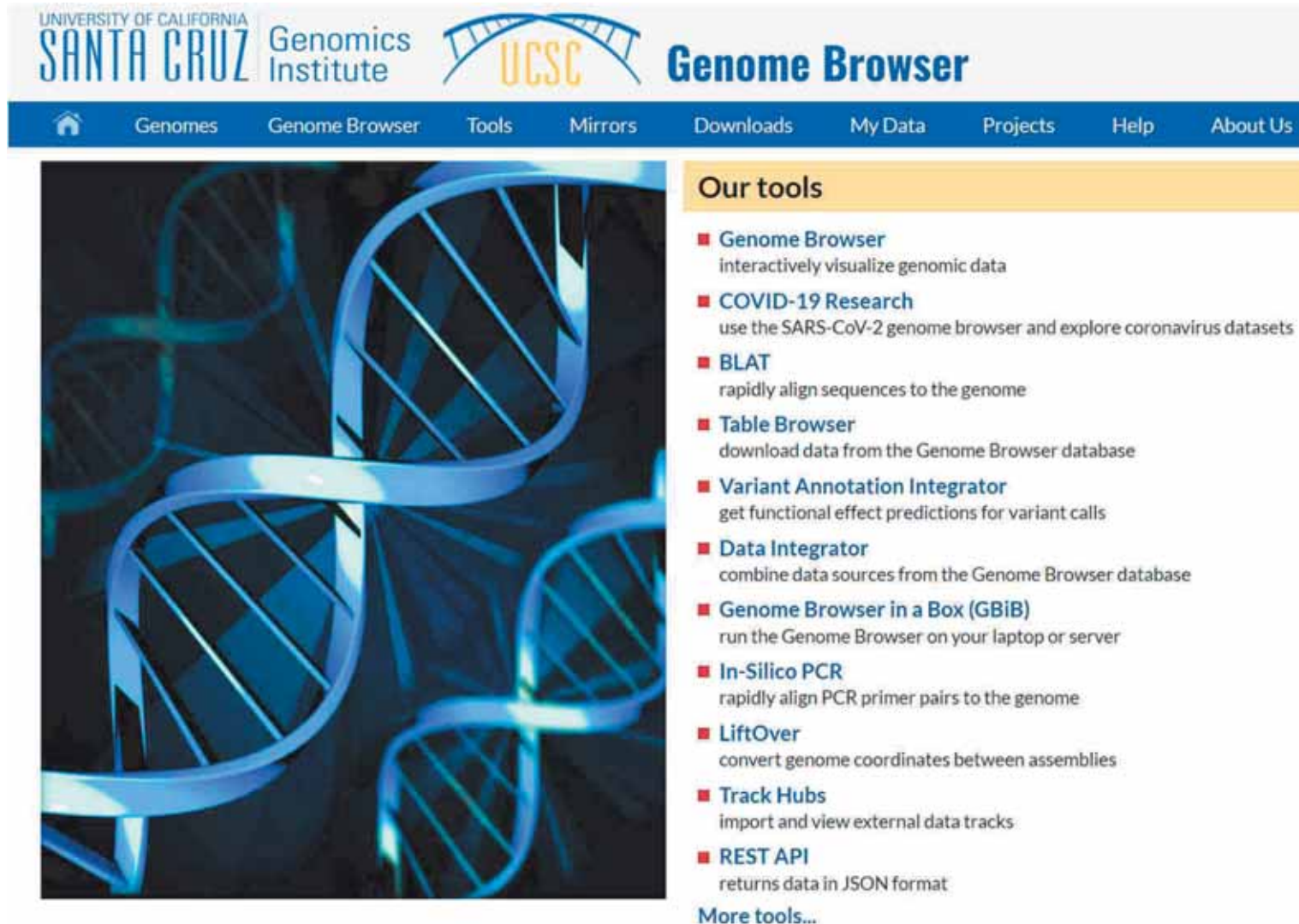
- イントロダクション
  - マイクロアレイ染色体検査 (CMA) 検査 超入門
  - BEDファイル形式データの取扱い
  - マイクロアレイ染色体検査の結果解釈の補助ソフトウェアツール (CAS) の使用方法
  - マイクロアレイ (CMA) 検査の結果解釈に必要なデータベースの使用法
- 実践編
  - 解析例 1 : CASを使用して疾患関連性を調べたLossの例
  - 解析例 2 : CASと各種データベースを使用して疾患関連性を調べたLossの例
  - 解析例 3 : CASと各種データベースを使用して疾患関連性を調べたGainの例
  - 解析例 4 : 疾患関連性領域近傍にあるBenignのLossの例
- Advanced編
  - データベース「UCSC」使用方法
  - 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

この動画ではマイクロアレイ染色体検査（以下、CMA）の「結果を見る」「結果を解釈する」ために必要な最低限の基本事項を述べています。

データベースの使用方法は様々なアレンジがあります。実践しながらオリジナルの使用方法を確立していただければと思います。

# UCSC Genome Browser

University of California  
SANTA CRUZ



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SANTA CRUZ Genomics Institute UCSC Genome Browser

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## Our tools

- **Genome Browser**  
interactively visualize genomic data
- **COVID-19 Research**  
use the SARS-CoV-2 genome browser and explore coronavirus datasets
- **BLAT**  
rapidly align sequences to the genome
- **Table Browser**  
download data from the Genome Browser database
- **Variant Annotation Integrator**  
get functional effect predictions for variant calls
- **Data Integrator**  
combine data sources from the Genome Browser database
- **Genome Browser in a Box (GBiB)**  
run the Genome Browser on your laptop or server
- **In-Silico PCR**  
rapidly align PCR primer pairs to the genome
- **LiftOver**  
convert genome coordinates between assemblies
- **Track Hubs**  
import and view external data tracks
- **REST API**  
returns data in JSON format

[More tools...](#)

<http://www.genome.ucsc.edu/> (2022.5.7アクセス)

# UCSC Genome Browser使用の流れ

## 1. Custom TracksにCNVデータを入力

CNV領域が表示される

- ✓ Zoom Outして領域全体を見渡しやすくする
- ✓ HighlightでCNV領域を見やすくする

## 2. 必要な情報表示の選択をする

CNVの病原性評価に有用な表示情報(一例)

- ✓ Base position, Chromosome Band
- ✓ ClinGen (HI/TS Score)
- ✓ ClinGen CNVs (Curated Pathogenic/Benign)
- ✓ DGV Gold Standard (一般集団CNV→”Benign示唆”)

## 3. 表示内容から病原性を判断する

※具体的な使用は症例解析の配信を参照ください。

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Our tools

- Genome Browser interactively visualize genomic data
- COVID-19 Research use the SARS-CoV-2 genome and coronavirus datasets
- BLAT rapidly align sequences to the genome
- Table Browser download data from the Genome Browser database
- Variant Annotation Integrator get functional effect predictions for variant calls
- Data Integrator combine data sources from the Genome Browser database

Custom Tracks My Sessions Track Hubs Track Collection Builder Public Sessions

クリック

- ① Home画面のMy Dataにカーソルを合わせる
- ② ドロップダウンリスト中のCustom Tracksをクリック

BEDファイルの

“22 18912231 21465672 Loss”を病原性評価



①

Display your own data as custom annotation tracks in the browser. Data must be formatted in [bigBed](#), [bigBarChart](#), [bigChain](#), [bigGenePred](#), [bigInteract](#), [bigLolly](#), [bigMaf](#), [bigPsl](#), [bigWig](#), [BAM](#), [barChart](#), [VCF](#), [BED](#), [BED detail](#), [bedGraph](#), [broadPeak](#), [CRAM](#), [GFF](#), [GTF](#), [hic](#), [interact](#), [MAF](#), [narrowPeak](#), [Personal Genome SNP](#), [PSL](#), or [WIG](#) formats.

- You can paste just the URL to the file, without a "track" line, for bigBed, bigWig, bigGenePred, BAM and VCF.
- To configure the display, set [track](#) and [browser](#) line attributes as described in the [User's Guide](#).

Examples are [here](#). If you do not have web-accessible data storage available, please see the [Hosting](#) section of the Track Hub Help documentation.

Please note a much more efficient way to load data is to use [Track Hubs](#), which are loaded from the [Track Hubs Portal](#) found in the menu under [My Data](#).

Paste URLs or data: Or upload:  ファイルが選択されていません

②

track name=**任意の名称①** description="**任意の名称②**" color=255,0,0

#chrom chromStart chromEnd  
22 18912231 21465672

染色体番号

始点のゲノムポジション

終点のゲノムポジション

色の例  
赤: 255,0,0  
青: 0,0,255  
黒: 0,0,0

③

クリック

① 赤枠内で「Mammal」「Human」「GRCh37/hg19」を選択

② Paste URLs or dataの下の空白に必要事項を記入

③ Submitをクリック



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### Manage Custom Tracks

genome: Human assembly: Feb. 2009 (GRCh37/hg19) [hg19]

Name	Description	Type	Doc	Items	Pos	delete
22q11.2	"22q11.2	bed		1	chr22:	<input type="checkbox"/>

view in

クリック

「go」をクリックすると結果が表示される



UCSC Genome Browser on Human (GRCh37/hg19)

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

multi-region chr22:18,912,232-21,465,672 2,553,441 bp.   [examples](#)

chr22 (q11.21) 22p13 22p12 22p11.2 22q11.21 q11.23 22q12.1 22q12.2 22q12.3 22q13.1 22q13.2 22q13.31 q13.33

Scale chr22: 19,500,000 | 1 Mb | 20,000,000 | 20,500,000 | hg19 | 21,000,000

Chromosome Bands Localized by FISH Mapping Clones 22q11.21

NCBI RefSeq genes, curated subset (NM\_\*, NR\_\*, NP\_\*, YP\_\*) - Annotation Release NCBI Homo sapiens 105.20220307 (2022-03-12)

PRODH/NM\_016335.6  
RODH/NM\_001195226.2  
22455341/NR\_173080.1  
22455341/NR\_173081.1  
DGCR5/NR\_024159.2  
DGCR5/NR\_026651.2  
DGCR5/NR\_110533.2  
DGCR5/NR\_045121.2  
DGCR5/NR\_002733.3  
1246C/NM\_001396027.1  
FAM246C/NR\_173148.1  
DGCR2/NM\_005137.3  
GCR2/NM\_001184781.2  
GCR2/NM\_001173533.2  
GCR2/NM\_001173534.2  
DGCR2/NR\_033674.2  
DGCR11/NR\_024157.1  
ESS2/NM\_022719.3  
ESS2/NR\_134304.2  
TSSK2/NM\_053006.5  
GSC2/NM\_005315.2  
INC01311/NR\_103767.1  
225A1/NM\_001256534.2  
225A1/NM\_001287387.2

HIRA/NM\_003325.4  
MRPL40/NM\_003776.4  
MRPL40/NM\_001318151.2  
C22orf39/NM\_173793.5  
C22orf39/NM\_001166242.2  
UFD1/NM\_001362910.2  
UFD1/NM\_005659.7  
UFD1/NM\_001035247.3  
CDC45/NM\_001369291.1  
CDC45/NM\_001178011.2  
CDC45/NM\_001178010.2  
CDC45/NM\_003504.5  
CDC45/NR\_161281.1  
CLDN5/NM\_001130861.1  
CLDN5/NM\_001363086.2  
CLDN5/NM\_001363067.2  
CLDN5/NM\_003277.4  
LINC00895/NR\_024381.1  
SEPTIN5/NM\_002688.6  
SEPT5-GP1BB/NR\_037611.1  
SEPTIN5/NM\_001009939.3  
SEPT5-GP1BB/NR\_037612.1  
GP1BB/NM\_000407.5  
MIR185/NR\_029706.1  
DGCR8/NM\_001190326.2  
TRMT2A/NM\_001331039.2  
TRMT2A/NM\_001257994.2  
TRMT2A/NM\_162984.5  
MIR4761/NR\_039918.1

RTN4R/NM\_023004.6  
MIR1286/NR\_011618.1  
DGCR6L/NM\_033257.4  
FAM230A/NR\_165629.1  
FAM230A/NR\_136560.2  
GGTLC3/NM\_001355479.1  
TMEM191B/NM\_001242313.1  
FAM230G/NR\_136572.2  
ZNF74/NR\_001256524.2  
ZNF74/NR\_001256523.2  
SCARF2/NM\_153334.7  
SCARF2/NM\_182895.5  
KLHL22/NR\_033825.2  
MED15/NM\_001293235.2  
MED15/NM\_001293236.2  
MED15/NM\_001293234.2  
MED15/NM\_015899.5  
MED15/NM\_001003891.3  
MED15/NM\_001293237.2

Pi4KA/NM\_058004.4  
SERPIND1/NM\_000185.4  
SNAP29/NM\_004782.4  
POM121L4P/NR\_024592.1  
TMEM191A/NR\_025815.1  
Pi4KA/NM\_001362862.2  
Pi4KA/NM\_001362863.2  
CRKL/NM\_005207.4  
CRKL/NR\_156180.2  
LINC01637/NR\_110537.1  
AIFM3/NM\_001018060.3  
AIFM3/NM\_144704.3  
AIFM3/NM\_001386814.1  
AIFM3/NM\_001146288.2  
AIFM3/NR\_027464.2  
THAP7/NM\_001008695.1  
THAP7-AS1/NR\_027052.1  
THAP7-AS1/NR\_027051.1  
TUBA3FP/NR\_003608.1  
P2RX6/NM\_001394694.1  
P2RX6/NM\_001394695.1  
P2RX6/NM\_001159554.2  
P2RX6/NM\_001349875.2  
P2RX6/NM\_001394693.1

入力したCustom Tracksの領域

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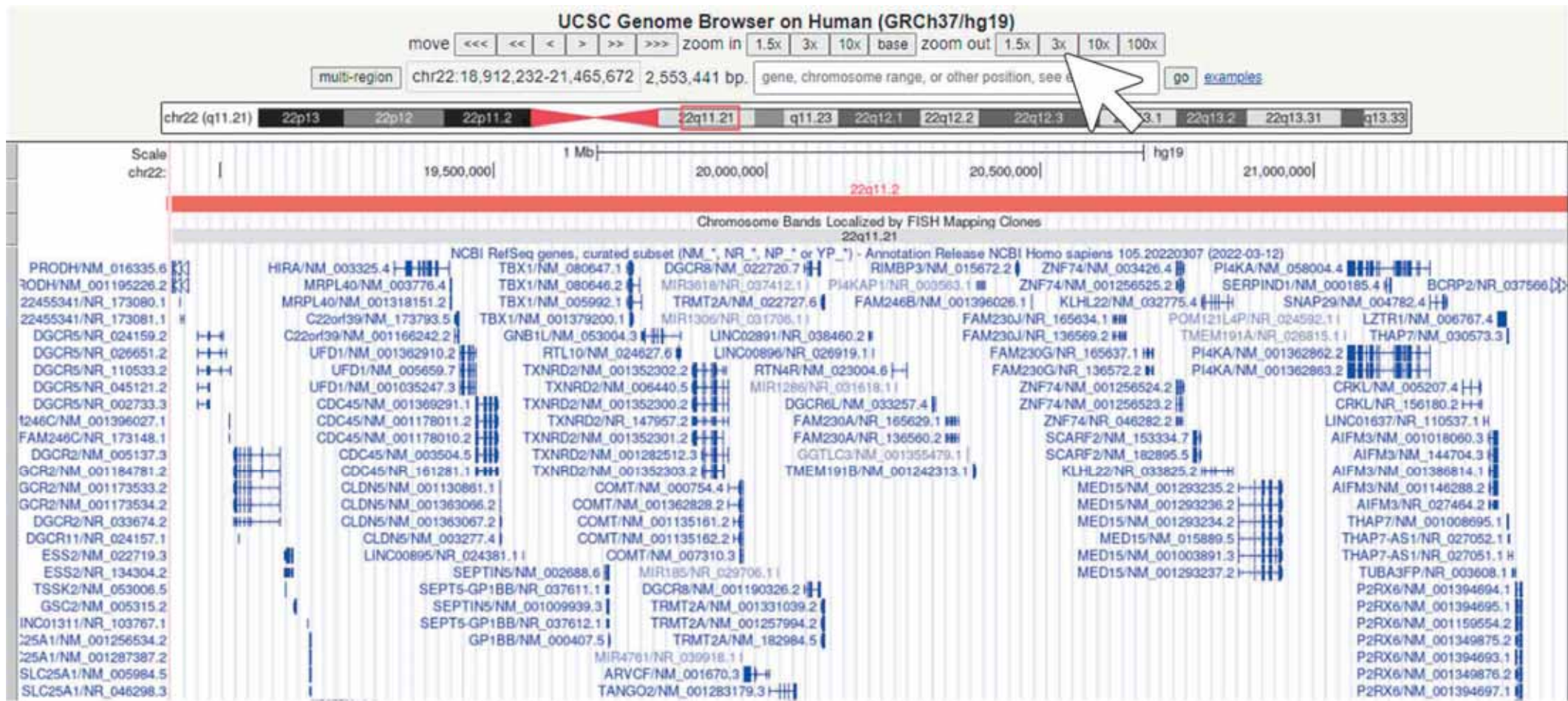
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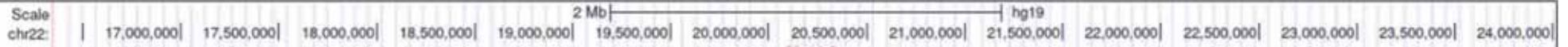


Zoom out 「3 × 」ないし「10 × 」をクリックして全体像をみる

### UCSC Genome Browser on Human (GRCh37/hg19)

move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x

multi-region chr22:16,358,791-24,019,113 7,660,323 bp, gene, chromosome range, or other position, see examples go [examples](#)



#### Chromosome Bands Localized by FISH Mapping Clones



NCBI RefSeq genes, curated subset (NM, \*, NR, \*, NP, \* or YP, \*) - Annotation Release NCBI Homo sapiens 105.20220307 (2022-03-12)

111H1/NM_001005239.2	GAB4/NM_001037814.1	MIR648/NR_030378.1	HIRA/NM_003325.4	PI4KAP1/NR_003503.1	CRKL/NM_005207.4	PPM1F/NM_014634.4	MIR5571/NR_049835.1
CCT8L2/NM_014406.5	ATP6V1E1/NM_001696.4	DGCR2/NM_005137.3	MIR185/NR_029706.1	TMEM191A/NR_026815.1	YDJC/NR_163924.1	PRAME/NM_206955.3	PCAT14/NR_109832.1
TPTEP1/NR_001591.1	BCL2L13/NM_015367.4	ESS2/NM_022719.3	DGCR8/NM_022720.7	PI4KA/NM_058004.4	MAPK1/NM_002745.5	PRAME/NM_206955.3	MIR650/NR_030755.1
PARP4P3/NR_040115.1	BCL2L13/NR_073069.1	ESS2/NR_134304.2	MIR3618/NR_037412.1	PI4KA/NM_001362862.2	MAPK1/NM_138957.3	IGLL5/NM_001256296.2	
LINC01665/NR_134584.1	BID/NM_197966.3	TSSK2/NM_053006.5	MIR1306/NR_031706.1	PI4KA/NM_001362863.2	PPM1F-AS1/NR_147620.1	RSPH14/NM_014433.3	
XKR3/NM_001318251.3	BID/NM_001196.4	GSC2/NM_005315.2	TRMT2A/NM_022727.6	SERPIND1/NM_000185.4	YDJC/NR_163925.1	PRAME/NM_206956.3	IGLL1/NM_001369906.1
XKR3/NM_175878.5	BID/NM_001244572.1	CLTCL1/NM_007098.4	RTN4R/NM_023004.6	SNAP29/NM_004782.4	TOP3B/NM_003935.5	GNAZ/NM_002073.4	
XKR3/NM_001386955.1	BID/NM_001244569.1	CLTCL1/NM_001835.4	MIR1286/NR_031618.1	CRKL/NR_156180.2	TOP3B/NM_001349852.2	RAB36/NM_001349878.1	
XKR3/NM_001386957.1	BID/NM_197967.2	MRPL40/NM_003776.4	RIMBP3/NM_015672.2	AIFM3/NM_144704.3	TOP3B/NR_146277.2	RAB36/NM_001349877.1	
XKR3/NM_001386956.1	BID/NM_001244570.1	MRPL40/NM_001318151.2	FAM230J/NR_165634.1	GGT2P/NR_172944.1	VPREB1/NM_007128.4	RAB36/NM_004914.5	
HSFY1P1/NR_003607.1	BID/NM_001244567.1	C22orf39/NM_173793.5	FAM230I/NR_136569.2	FAM230H/NR_136559.2	BMS1P20/NR_027293.2	BCR/NM_021574.3	
GAB4/NM_001386957.1	LINC01634/NR_024417.1	UFD1/NM_005659.7	FAM230G/NR_165637.1	LINC01651/NR_170329.1	ZNF280B/NM_080764.4	FAM230I/NR_165490.1	
GAB4/NR_159481.1	PEX26/NM_001199319.2	CDC45/NM_003504.5	FAM230G/NR_136572.2	HIC2/NM_015094.3	ZNF280B/NR_130642.2	FAM230I/NR_110539.2	
CECR7/NR_152826.1	PEX26/NM_017929.6	CDC45/NR_161281.1	ZNF74/NM_001256525.2	PIAKAP2/NR_003700.1	ZNF280B/NR_130643.2	FAM230I/NR_165489.1	
CECR7/NR_152825.1	PEX26/NM_001127649.3	CLDN5/NM_003277.4	ZNF74/NM_001256524.2	RIMBP3C/NM_001128633.2	ZNF280A/NM_080740.5	FAM230I/NR_165488.1	
CECR7/NR_015352.2	TUBA8/NM_001193414.2	SEPTIN5/NM_002688.6	ZNF74/NM_001256523.2	UBE2L3/NM_001256355.1	PRAME/NM_206953.3	IGLL1/NM_020070.4	
IL17RA/NM_001289905.2	TUBA8/NM_018943.3	SEPTIN5/NM_001009939.3	ZNF74/NM_003426.4	UBE2L3/NM_001256356.2	PRAME/NM_006115.5	IGLL1/NM_152855.3	
IL17RA/NM_014339.7	USP18/NM_017414.4	GP1BB/NM_000407.5	ZNF74/NR_046282.2	UBE2L3/NM_003347.4	PRAME/NM_206954.3	DRICH1/NM_016449.4	
TMEM121B/NM_031890.4	FAM230D/NR_136570.2	TBX1/NM_080647.1	SCARF2/NM_153334.7	UBE2L3/NR_028436.3	GGTLC2/NM_199127.3	GUSBP11/NR_024448.3	
TMEM121B/NM_001163079.2	FAM230E/NR_136561.2	TBX1/NM_080646.2	SCARF2/NM_182895.5	UBE2L3/NR_046082.2	IGLL5/NM_001178126.2		
LINC01664/NR_103793.1	FAM230E/NR_165635.1	TBX1/NM_005992.1	KLHL22/NM_032775.4	YDJC/NM_001017964.2	RAB36/NR_146295.3		
HDHD5/NM_033070.3	GGT3P/NR_003267.1	TBX1/NM_001379200.1	KLHL22/NR_033825.2	YDJC/NM_001371350.1	BCR/NM_004327.4		
HDHD5/NM_017829.6	POM121L15P/NR_170942.1	GNB1L/NM_053004.3	MED15/NM_001293235.2	YDJC/NR_163922.1	FBXW4P1/NR_033408.1		
HDHD5-AS1/NR_024482.1	POM121L15P/NR_135922.1	RTL10/NM_024627.6	MED15/NM_001293236.2	YDJC/NR_163923.1	LINC02556/NR_149133.1		
HDHD5-AS1/NR_024483.1	FAM230F/NR_136571.2	TXNRD2/NM_006440.5	MED15/NM_015889.5	CCDC116/NM_152612.3	CESSAP1/NR_037839.1		
ADA2/NM_001282225.2	DGCR6/NM_005675.6	TXNRD2/NR_147957.2	POM121L4P/NR_024592.1	SDF2L1/NM_022044.3	ZDHHC8P1/NR_003950.1		
ADA2/NM_001282226.2	PRODH/NM_016335.6	COMT/NM_000754.4	LINC01637/NR_110637.1	TOP3B/NM_001349850.2	LINC01650/NR_110538.1		
ADA2/NM_001282228.2	PRODH/NM_001195226.2	COMT/NM_001362828.2	AIFM3/NM_001018060.3	PRAMENP/NR_135291.1			
ADA2/NM_001282229.2	LOC122455341/NR_173080.1	COMT/NM_001135161.2	AIFM3/NM_001386814.1	VPREB1/NM_001303509.2			
ADA2/NM_001282227.2	LOC122455341/NR_173081.1	COMT/NM_001135162.2	AIFM3/NM_001146288.2	PRAME/NM_001318127.2			
ADA2/NM_177405.3	DGCR5/NR_024159.2	COMT/NM_007310.3	AIFM3/NR_027464.2	PRAME/NM_001318126.2			
CECR3/NR_038398.2	DGCR5/NR_026651.2	MIR4761/NR_039918.1	LZTR1/NM_006767.4	PRAME/NM_001291715.2			
CECR2/NM_001290046.2	DGCR5/NR_110533.2	ARVCF/NM_001670.3	THAP7/NM_030573.3	PRAME/NM_001291716.2			
CECR2/NM_001290047.2	DGCR5/NR_045121.2	TANGO2/NR_136212.1	THAP7/NM_001008695.1	PRAME/NM_001291719.2			
SLC25A18/NM_001303484.2	DGCR5/NR_002733.3	TANGO2/NM_152906.7	THAP7-AS1/NR_027052.1	PRAME/NM_001291717.2			
SLC25A18/NM_031481.3	DGCR2/NR_033674.2	TRMT2A/NM_182984.5	THAP7-AS1/NR_027051.1	LL22NC03-63E9.3/NR_027426.2			
LOC101929372/NR_171783.1	FAM246C/NR_173148.1	TANGO2/NR_104274.3	TUBA3FP/NR_003608.1	POM121L1P/NR_024591.1			
ATP6V1E1/NM_001039367.1	DGCR11/NR_024157.1	TANGO2/NR_104275.3	P2RX6/NM_001394694.1	GGTLC2/NM_001282879.2			
ATP6V1E1/NM_001039366.1	LINC01311/NR_103767.1	MIR6816/NR_106874.1	P2RX6/NM_001394695.1	GGTLC2/NM_001391911.1			
BCL2L13/NM_001363824.1	SLC25A1/NM_005984.5	RANBP1/NM_002882.4	P2RX6/NM_001159554.2	GGTLC2/NM_001391910.1			
BCL2L13/NM_001270727.1	SLC25A1/NR_046298.3	ZDHHC8/NM_013373.4	P2RX6/NM_001349875.2				

UCSC Genome Browser on Human (GRCh37/hg19)

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multi-region chr22:16,358,791-24,019,113 7,660,323 bp. gene, chromosome range, or other position, see examples go examples

chr22 (q11.1-q11.23) 22p13 22p12 22p11.2 22q11.21 22q11.23 22q12.1 22q12.2 22q12.3 22q13.1 22q13.2 22q13.31 22q13.33

Scale chr22: | 17,000,000 | 17,500,000 | 18,000,000 | 18,500,000 | 19,000,000 | 19,500,000 | 20,000,000 | 20,500,000 | 21,000,000 | 21,500,000 | 22,000,000 | 22,500,000 | 23,000,000 | 23,500,000 | 24,000,000 | hg19

2 Mb

Chromosome Browser visualized by FeatureTrack

hide  
dense  
squish  
✓ pack  
full  
Zoom to feature  
Highlight feature  
Get DNA features  
Open details in new window...  
Show details for feature...  
Configure 22q11.2  
View image

①

②

NCBI RefSeq genes, curated subset (NM \*, NR \*, NP \*, etc.) Annotatic

GAB4/NM\_001037814.1 | MIR648/NR\_030378.1 | HIRA/NM\_003325.4 | PI4KAP1/NR\_015513.1 |

ATP6V1E1/NM\_001696.4 | DGCR2/NM\_005137.3 | MIR185/NR\_029706.1 |

BCL2L13/NM\_015367.4 | ESS2/NM\_022719.3 | DGCR8/NM\_022720.7 |

BCL2L13/NR\_073069.1 | ESS2/NR\_134304.2 | MIR3618/NR\_037412.1 |

BID/NM\_197968.3 | TSSK2/NM\_053008.5 | MIR1306/NR\_031706.1 |

BID/NM\_001196.4 | GSC2/NM\_005315.2 | TRMT2A/NM\_022727.6 | SERP1

BID/NM\_001244572.1 | CLTCL1/NM\_007098.4 | RTN4R/NM\_023004.6 | S

BID/NM\_197967.2 | MRPL40/NM\_003776.4 | RIMBP3/NM\_015672.2 |

BID/NM\_001244570.1 | MRPL40/NM\_001318151.2 | FAM230J/NR\_1656

HSFY1P1/NR\_003607.1 | BID/NM\_001244567.1 | C22orf39/NM\_173793.5 | FAM230J/NR\_1365

GAB4/NM\_001366957.1 | LINC01634/NR\_024417.1 | UFD1/NM\_005659.7 |

GAB4/NR\_159481.1 | PEX26/NM\_001199319.2 | CDC45/NM\_003504.5 |

CECR7/NR\_152826.1 | PEX26/NM\_017929.6 | CDC45/NR\_161281.1 |

CECR7/NR\_152825.1 | PEX26/NM\_001127649.3 | CLDN5/NM\_003277.4 |

CECR7/NR\_015352.2 | TUBA8/NM\_001193414.2 | SEPTIN5/NM\_002688.6 |

IL17RA/NM\_001289905.2 | TUBA8/NM\_018943.3 | SEPTIN5/NM\_001009939.3 |

IL17RA/NM\_014339.7 | USP18/NM\_017414.4 | GP1BB/NM\_000407.5 |

TMEM121B/NM\_031890.4 | FAM230D/NR\_136570.2 | TBX1/NM\_080647.1 |

TMEM121B/NM\_001163079.2 | FAM230E/NR\_136561.2 | TBX1/NM\_080646.2 |

LINC01664/NR\_103793.1 | FAM230E/NR\_165635.1 | TBX1/NM\_005992.1 |

HDHD5/NM\_033070.3 | GGT3P/NR\_003267.1 | TBX1/NM\_001379200.1 |

HDHD5/NM\_017829.6 | POM121L15P/NR\_170942.1 | GNB1L/NM\_053004.3 |

HDHD5-AS1/NR\_024482.1 | POM121L15P/NR\_135922.1 | RTL10/NM\_024627.6 |

ADA2/NM\_001282225.2 | FAM230F/NR\_136571.2 | TXNRD2/NM\_006440.5 |

ADA2/NM\_001282226.2 | DGCR6/NM\_005675.6 | TXNRD2/NR\_147957.2 |

PRODH/NM\_016335.6 | COMT/NM\_000754.4 | LINC01637/NR\_110537.1 | TOP3B/NM\_001349850.2 |

LINC01665/NR\_134584.1 |

XKR3/NM\_001318251.3 |

XKR3/NM\_175878.5 |

XKR3/NM\_001386955.1 |

XKR3/NM\_001386957.1 |

XKR3/NM\_001386956.1 |

LINC01665/NR\_134584.1 |

BID/NM\_197968.3 |

BID/NM\_001196.4 |

BID/NM\_001244572.1 |

BID/NM\_197967.2 |

BID/NM\_001244570.1 |

HSFY1P1/NR\_003607.1 |

GAB4/NM\_001366957.1 |

GAB4/NR\_159481.1 |

CECR7/NR\_152826.1 |

CECR7/NR\_152825.1 |

CECR7/NR\_015352.2 |

IL17RA/NM\_001289905.2 |

IL17RA/NM\_014339.7 |

TMEM121B/NM\_031890.4 |

TMEM121B/NM\_001163079.2 |

LINC01664/NR\_103793.1 |

HDHD5/NM\_033070.3 |

HDHD5/NM\_017829.6 |

HDHD5-AS1/NR\_024482.1 |

ADA2/NM\_001282225.2 |

ADA2/NM\_001282226.2 |

PRODH/NM\_016335.6 |

COMT/NM\_000754.4 |

LINC01637/NR\_110537.1 |

TOP3B/NM\_001349850.2 |

0307 (2022-03-12)

4.11 PRAME/NM\_206955.3 | PCAT14/NR\_109832.1 |

2745.5 | MIR650/NR\_030755.1 |

18957.3 | IGLL5/NM\_001256296.2 |

R\_147620.1 | RSPH14/NM\_014433.3 |

5.11 PRAME/NM\_206956.3 | IGLL1/NM\_001369906.1 |

M\_003935.5 | GNAZ/NM\_002073.4 |

001349852.2 | RAB36/NM\_001349878.1 |

NR\_146277.2 | RAB36/NM\_001349877.1 |

?REB1/NM\_007128.4 | RAB36/NM\_004914.5 |

MS1P20/NR\_027293.2 | BCR/NM\_021574.3 |

ZNF280B/NM\_080764.4 | FAM230I/NR\_165490.1 |

ZNF280B/NR\_130642.2 | FAM230I/NR\_110539.2 |

ZNF280B/NR\_130643.2 | FAM230I/NR\_165489.1 |

ZNF280A/NM\_080740.5 | FAM230I/NR\_165488.1 |

PRAME/NM\_206953.3 | IGLL1/NM\_020070.4 |

PRAME/NM\_006115.5 | IGLL1/NM\_152855.3 |

PRAME/NM\_206954.3 | DRICH1/NM\_016449.4 |

GGTLC2/NM\_199127.3 | GUSBP11/NR\_024448.1 |

IGLL5/NM\_001178126.2 |

RAB36/NR\_146295.3 |

BCR/NM\_004327.4 |

FBXW4P1/NR\_033408.1 |

LINC02556/NR\_149133.1 |

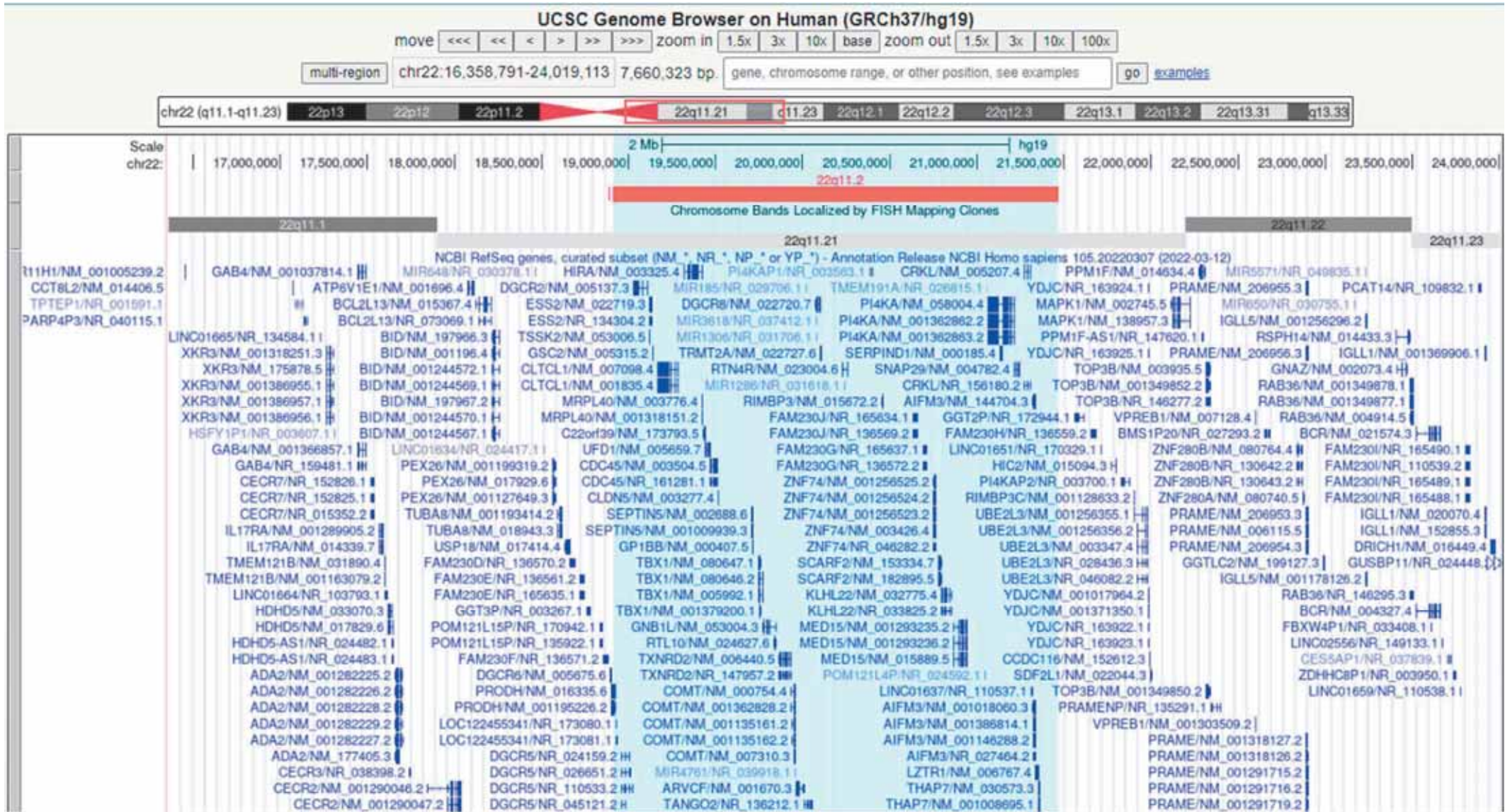
CESSAP1/NR\_037839.1 |

ZDHHC8P1/NR\_003950.1 |

LINC01659/NR\_110538.1 |

① Custom Tracksで表示したバーの上で右クリック

② リスト中のHighlight featureをクリック



Custom Tracksの領域が強調される

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- ✓ ClinGen CNVs (Curated Pathogenic/Benign)
- ✓ DGV Gold Standard (一般集団CNV→”Benign示唆”)

## 3. 表示内容から病原性を判断する

# 画面下方の各種データの表示・非表示を選択していく

track search | default tracks | default order | hide all | manage custom tracks | track hubs | configure | reverse | resize | refresh

collapse all | Use drop-down controls below and press refresh to alter tracks displayed. Tracks with lots of items will automatically be displayed in more compact modes. | expand all

**Custom Tracks** refresh

22q11.2 pack

**Mapping and Sequencing** refresh

Base\_Position | deCODE Recomb | Gap | Hi Sen Death | Problematic Regions | P12 Fix Patches | ENCODE Pilot | GC Percent | INSDC | Recomb Rate | P12 Alt Haplotypes | ENCODE Pilot | GRC Incident | jInOver & ReMap | Recomb Rate | RefSeq Acc | Assembly | Exome Probesets | GRC Map Contigs | Hg18 Diff | Map Contigs | Restr. Enzymes | BAC End Pairs | FISH Clones | Fosmid End Pairs | Hg38 Diff | Mappability | STS Markers | BL ORChID | FOSMID

**Genes and Gene Predictions** refresh

UCSC Genes | Exoniphy | MGC Genes | Retrospliced Genes | Vega Genes | NCBI RefSeq | GENCODE | Old UCSC Genes | snomiRNA | ORFeome Clones | TransMap V0 | CCDS | H-Iny 7.0 | CRISPR Targets | IKMC Genes Mapped | Other RefSeq | Pfam in UCSC Gene | Ensembl Genes | lincRNAs | Prediction Archive | UniProt | EvoFold | LRG Transcripts

**Phenotype and Literature** refresh

Publications | COSMIC Regions | Gene Interactions | LOVD Variants | REVEL Scores | Web Sequences | CADD | DECIPHER CNVs | GeneReviews | MGI Mouse OTL | RGD Human OTL | ClinGen | DECIPHER SNVs | GWAS Catalog | OMIM Alleles | RGD Rat OTL | ClinVar Variants | GAD View | HGMD Variants | OMIM Cytb Loci | SNpedia | Coriell CNVs | GenCC | Lens Patents | Orphanet | Variants in Papers

**COVID-19** refresh

COVID GWAS v1 | COVID GWAS v3 | Rare Harmful Vars

**mRNA and EST** refresh

CGAP SAGE | Other ESTs | UniGene | Gene Bounds | Other mRNAs | H-Iny | Poly(A) | Human ESTs | PolyA-Seq | Human mRNAs | SIR Alt-Splicing | Human RNA Editing | Spliced ESTs

**Expression** refresh

GTFx Gene V8 | GTFx RNA-seq | GTFx Transcript | Seston Brain | Allen Brain | EPDnew Promoters | GWIPS-viz Riboseq | Burge RNA-seq | Affy Archive | Illumina WG-6 | CSHL Small RNA-seq | ENC Exon Array | ENC ProtGeno | ENC RNA-seq | GIS RNA PET | PeptideAtlas | GNF Atlas 2 | gPCR Primers | RIKEN CAGE LOC | GTFx Gene

**Regulation** refresh

ENCODERegulation | ENC Histone | GTFx Combined eQTL | CHIP-seq | UCSF Brain Methyl | CD34-DnaseI | ENC RNA Binding | GTFx Tissue eQTL | Start Nucleosome | UMMS Brain Hist | CpG Islands | ENC TF Binding | JASPAR Transcription Factors | SUNY SwitchGear | UJW Repli-seq | VISTA Enhancers | ENC Chromatin | ENC DNA Methyl | GeneHancer | ORRegAnno | SwitchGear TSS | TFBS Conserved | ENC DNase/FAIRE | GenomE Segments | TS miRNA Targets

**Comparative Genomics** refresh

Conservation | Primate Chain/Net | Neandertal Assembly and Analysis | Denisova Assembly and Analysis | Variation | Repeats

Cons 46-Way | Cons Indels MmCf | Evo-Cpg | GERP | phastBias gBGC | Placental Chain/Net | Vertebrate Chain/Net | CHM13 alignments | 5% Lowest S | Cand\_Gene Flow | H-C Coding Diffs | Neandertal Methyl | Neandertal Mito [No data-chr22] | Neandertal Seq | S SNPs | Sel Swp Scan (S) | Mod Hum Variants | Modern Derived | gbSNP 153 | 1000G Archive | Array Probesets | gbSNP Archive | gbVar Common Struct Var | gGV Struct Var (pack) | EVS Variants | ExAC | Genome In a Bottle | Genome Variants | GIS DNA PET | gnomAD | HAIB Genotype | HapMap SNPs | HGDP Allele Freq | Platinum Genomes

**Repeats** refresh

RepeatMasker | Simple Repeats | Interspersed Repts | WM + SDust | Microsatellite | Numts Sequence | Segmental Dups | Self Chain

続き



## 表示選択項目の意味

---

- full: 情報一つに対して一行を使った表記  
縦幅をかなり使用する(スクロールが大変)
  - pack: 領域の重複がなければ情報を一行に複数表記  
fullより少し縦幅の節約ができる(スクロールが大変)
  - squish: 情報を示すバーを細くして表記  
かなり縦幅の節約ができる(意外と使いやすい)
  - dense: 情報の有無を一本のバーで表記  
縦幅ほとんど使わないが表示が見にくい
  - hide: 情報を表示しない
-

# UCSC Genome Browser使用の流れ

## 1. Custom TracksにCNVデータを入力

CNV領域が表示される

- ✓ Zoom Outして領域全体を見渡しやすくする
- ✓ HighlightでCNV領域を見やすくする

## 2. 必要な情報表示の選択をする

CNVの病原性評価に有用な表示情報(一例)

- ✓ Base position, Chromosome Band
- ✓ ClinGen (HI/TS Score)
- ✓ ClinGen CNVs (Curated Pathogenic/Benign)
- ✓ DGV Gold Standard (一般集団CNV→”Benign示唆”)

## 3. 表示内容から病原性を判断する

track search default tracks default order hide all manage custom tracks track hubs configure reverse resize refresh

collapse all Use drop-down controls below and press refresh to alter tracks displayed. Tracks with lots of items will automatically be displayed in more compact modes. expand all

Custom Tracks refresh

22q11.2 pack

①

Base Position dense

Chromosome Band pack

②

Mapping and Sequencing refresh

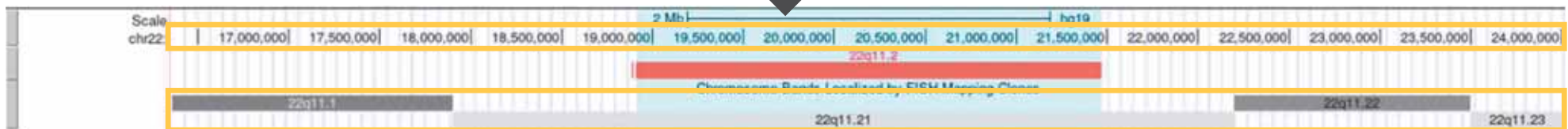
Genes and Gene Predictions refresh

下記の通りドロップダウンを選択

- ① Base Position : dense
- ② Chromosome Band : pack



Base Position



Chromosome Band

# UCSC Genome Browser使用の流れ

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## 3. 表示内容から病原性を判断する

Phenotype and Literature

ClinGen pack

①

ClinGen Track Settings

ClinGen curation activities (Dosage Sensitivity and Gene-Disease Validity)

Display mode: pack Submit Cancel Reset to defaults

Display data as a density graph:

List subtracks:  only selected/visible  all

②

Filter by Dosage Sensitivity Score (select multiple items - help)

Some evidence for dosage pathogenicity

Sufficient evidence for dosage pathogenicity

④

③

⑤

⑥

- ① ClinGenをクリック
- ② HIの選択をpack
- ③ TSの選択をpack
- ④ 「Filter by Dosage Sensitivity Score」下のカラムをクリック
- ⑤ ドロップダウンから下記選択
  - Some evidence... (HI Score: 2)
  - Sufficient evidence... (HI Score: 3)
- ⑥ 最後にSubmitをクリック

ClinGen Track Settings

ClinGen curation activities (Dosage Sensitivity and Gene-Disease Validity) (All Phenotype and Literature tracks)

Display mode: pack Submit Cancel Reset to defaults

Display data as a density graph:

List subtracks:  only selected/visible  all

⑥

Filter by Dosage Sensitivity Score (select multiple items - help)

Select multiple...

All

No evidence available

Little evidence for dosage pathogenicity

Some evidence for dosage pathogenicity

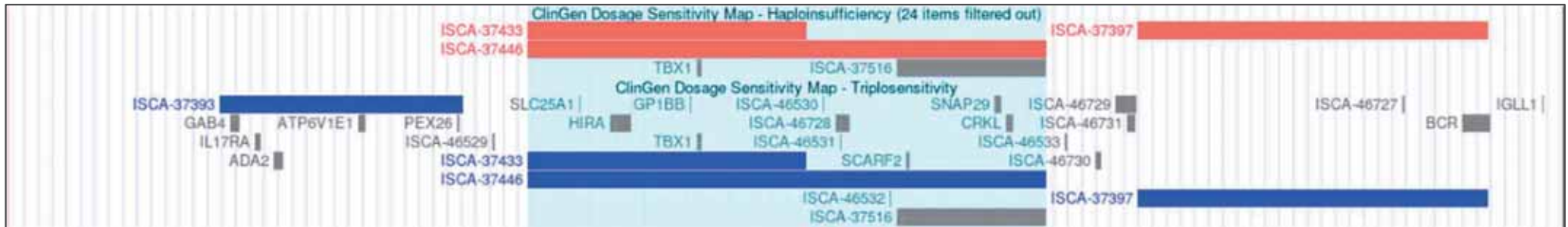
Sufficient evidence for dosage pathogenicity

Gene associated with autosomal recessive phenotype

Dosage sensitivity unlikely

close

⑤



- HI Score 3の領域および遺伝子 : 赤
- TS Score 3の領域および遺伝子 : 青
- HI/TS Score  $\leq 2$ の遺伝子および領域 : 灰色

# UCSC Genome Browser使用の流れ

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## 3. 表示内容から病原性を判断する

Phenotype and Literature

Publications CADD ClinGen CNVs ClinVar Variants Coriell CNVs  
hide hide pack full

COSMIC Regions DECIPHER CNVs DECIPHER SNVs Development Delay  
hide pack hide hide hide

Gene Interactions GeneReviews GWAS Catalog Haploinsufficiency HGM  
hide hide hide hide hide

LOVD Variants MGI Mouse OMIM Alleles OMIM Cyto Loci OMIM Genes  
hide QTL hide hide pack hide

REVEL Scores RGI ClinGen CNVs Track Settings  
hide QTL hide

Web Sequences

COVID GWAS COVID GWAS  
v4 v3  
hide hide

CGAP SAGE Gene Expression  
hide hide

Other ESTs Other ESTs  
hide hide

UniGene  
hide

Clinical Genome Resource (ClinGen) CNVs (\*All Phenotype and Literature tracks)

Maximum display mode: full Submit Cancel Reset to defaults

Select views (Help):  
Coverage: hide CNVs: pack

Coverage Configuration

Type of graph: bar Graph configuration help  
Track height: 57 pixels (range: 16 to 128)  
Data view scaling: use vertical viewing range setting Always include zero: ON  
Vertical viewing range: min: 0 max: 100 (range: 0 to 100)  
Transform function: Transform data points by: NONE  
Windowing function: mean+whiskers Smoothing window: OFF pixels  
Negate values:   
Draw y indicator lines: at y = 0.0 OFF at y = 0 OFF

Select subtracks by evidence and class:

All	Evidence	Curated	Submitted
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
<input type="checkbox"/>	<input type="checkbox"/>	<input checked="" type="checkbox"/>	<input type="checkbox"/>

List subtracks:  only selected/visible  all (3 of 11 selected)

	Class <sup>1</sup>	Evidence <sup>2</sup>	views <sup>3</sup>	Track Name <sup>4</sup>	Schema
<input checked="" type="checkbox"/> pack	Benign	Curated	CNVs	ClinGen CNVs: Curated Benign	Schema
<input type="checkbox"/> pack	Benign	Submitted	CNVs	ClinGen CNVs: Benign	Schema
<input type="checkbox"/> hide	Benign	Submitted	Coverage	ClinGen CNVs: Benign Gain Coverage	Schema
<input type="checkbox"/> hide	Benign	Submitted	Coverage	ClinGen CNVs: Benign Loss Coverage	Schema
<input type="checkbox"/> pack	Likely Benign	Submitted	CNVs	ClinGen CNVs: Uncertain: Likely Benign	Schema
<input type="checkbox"/> pack	Likely Pathogenic	Submitted	CNVs	ClinGen CNVs: Uncertain: Likely Pathogenic	Schema
<input checked="" type="checkbox"/> pack	Pathogenic	Curated	CNVs	ClinGen CNVs: Curated Pathogenic	Schema
<input type="checkbox"/> pack	Pathogenic	Submitted	CNVs	ClinGen CNVs: Pathogenic	Schema
<input type="checkbox"/> hide	Pathogenic	Submitted	Coverage	ClinGen CNVs: Pathogenic Gain Coverage	Schema
<input type="checkbox"/> hide	Pathogenic	Submitted	Coverage	ClinGen CNVs: Pathogenic Loss Coverage	Schema
<input checked="" type="checkbox"/> pack	Uncertain	Submitted	CNVs	ClinGen CNVs: Uncertain	Schema

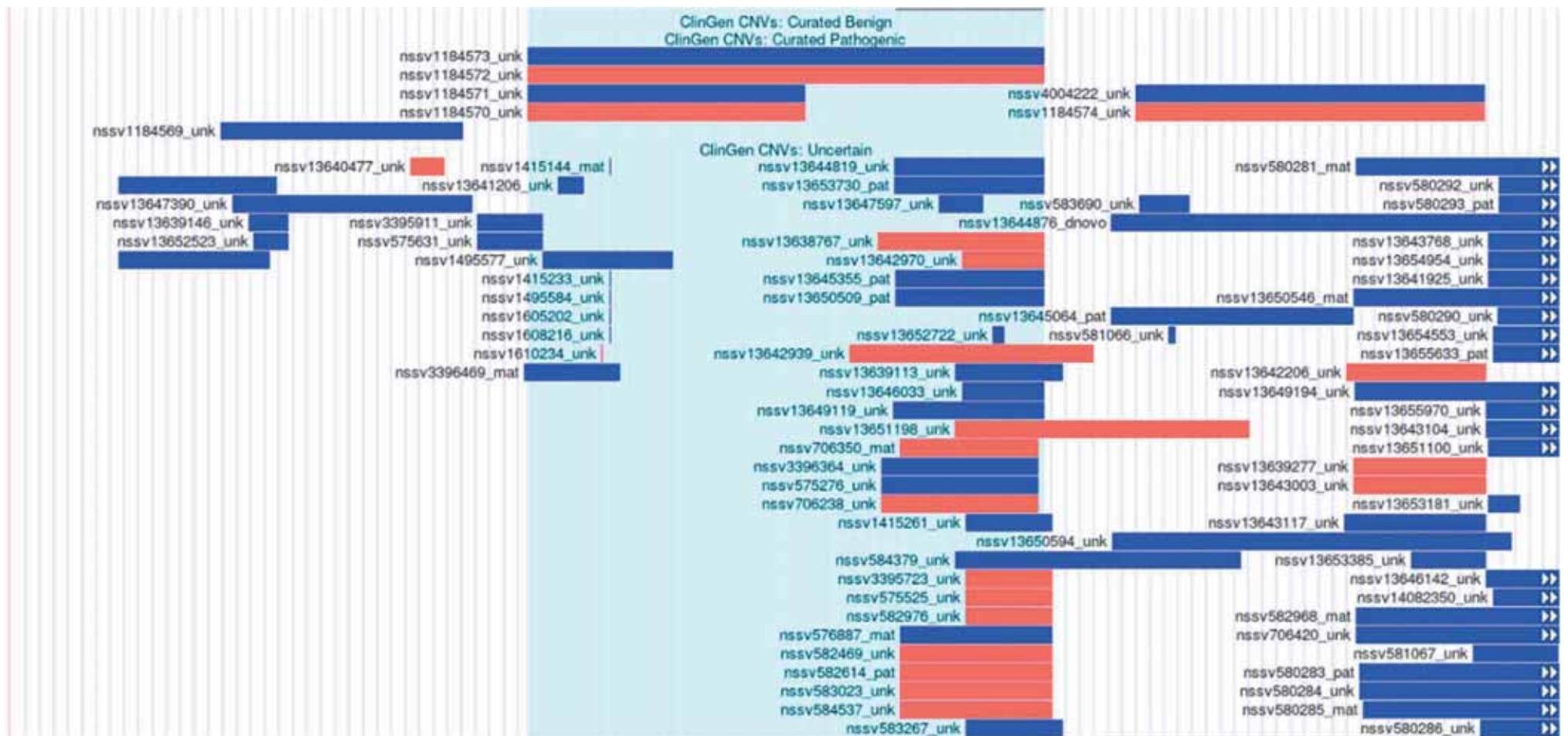
3 of 11 selected

Submit

- ① ClinGen CNVsをクリック
- ② Curated Benignをpack
- ③ Curated Pathogenicをpack
- ④ Uncertainをpack  
※入れなくともOK
- ⑥ 最後にSubmitをクリック

②③でBenign/Pathogenicが  
確立された領域を表示





- Curated Benignなし: 確立されたBenign CNVsなし
- Curated Pathogenicあり: 赤(欠失CNV)、青(重複CNV)
- Uncertain CNVs: 赤(欠失CNV)、青(重複CNV)

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## 3. 表示内容から病原性を判断する

# DGV

## Database of Genomic Variants

Database of Genomic Variants

*A curated catalogue of human genomic structural variation*

URL: <http://dgv.tcag.ca/dgv/app/home>

- 一般集団におけるCNVデータベース

- Structural Variants

一般集団で認められた様々なCNVを登録したもの

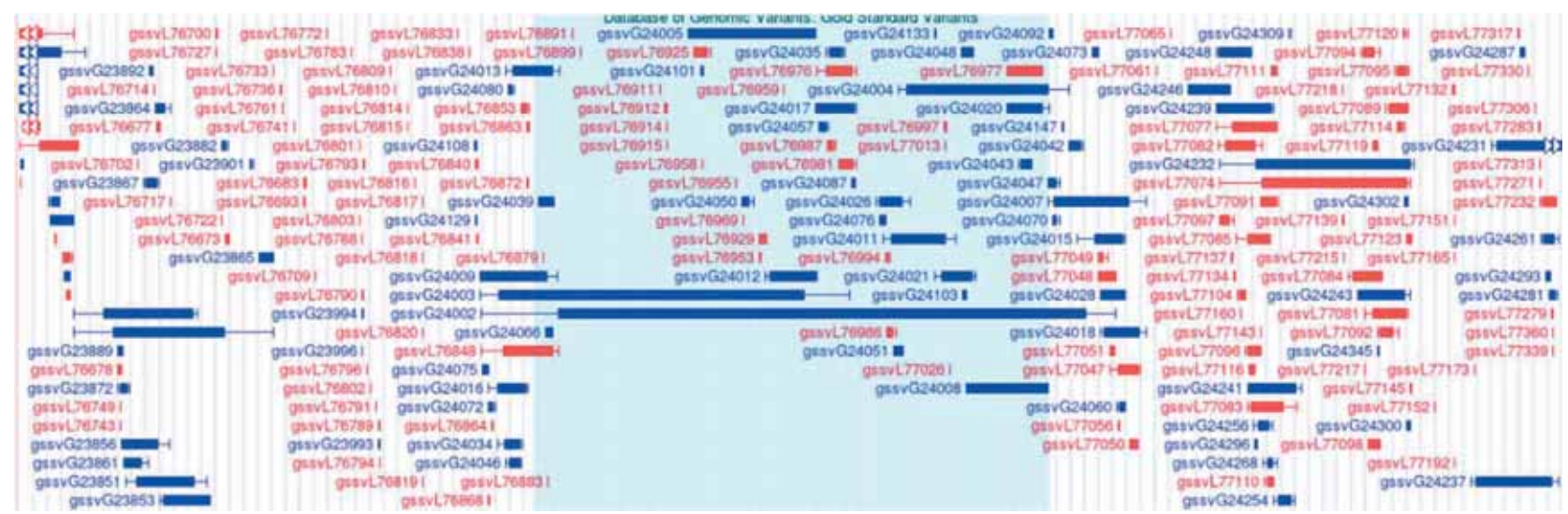
- Gold Standard Variants

しっかりと評価された一般集団研究に絞って認めら

れたCNVを登録したもの

Gold Standardに登録されたCNVsはBenignの可能性高い

- ① DGV Struct Varをクリック
- ② DGV Gold Standardをpack
- ③ 最後にSubmitをクリック



赤(欠失CNV) 青(重複CNV)

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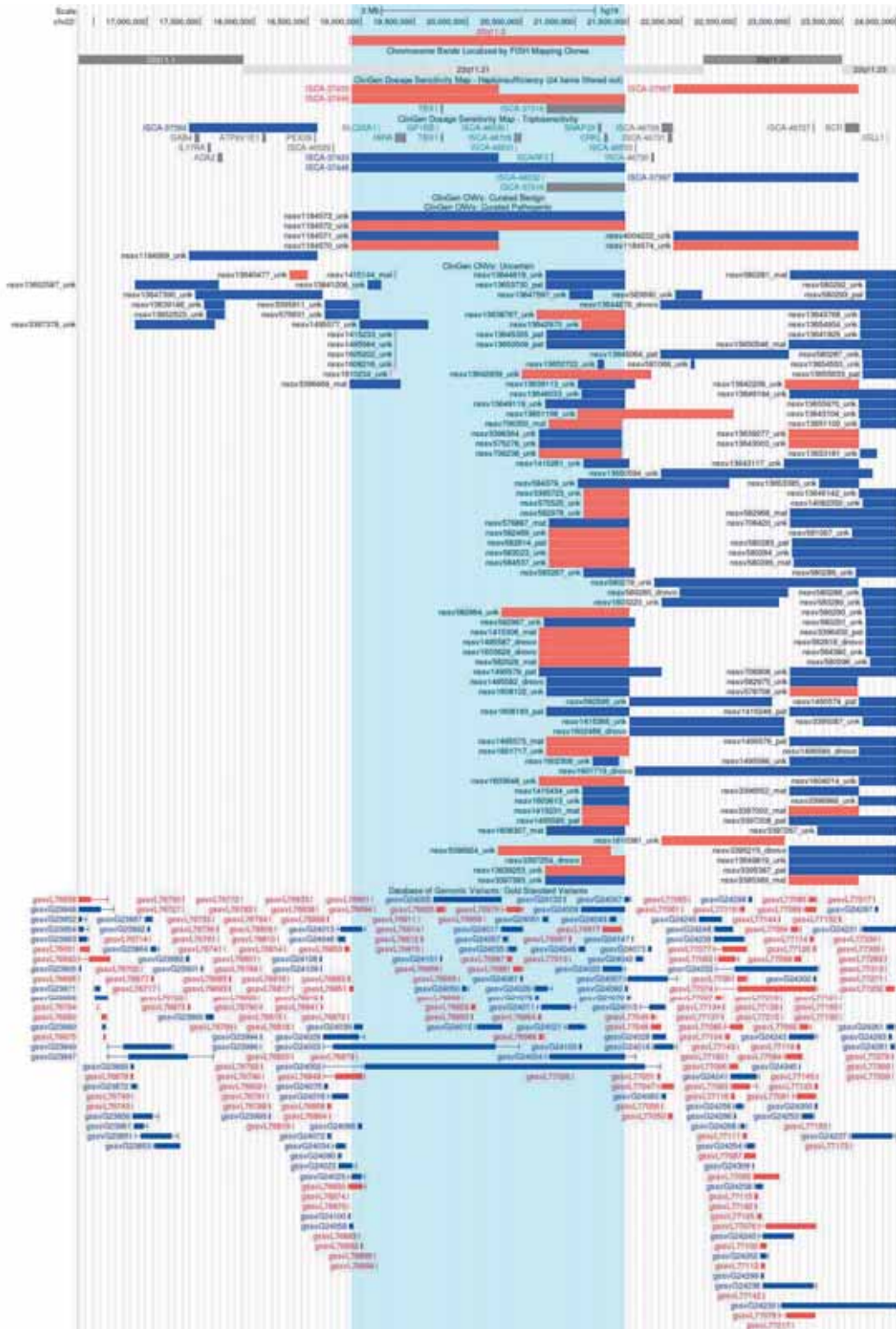
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## 3. 表示内容から病原性を判断する



## CNVの病原性評価

- HIが確立されている: HI Score 3  
TSも確立されている

- ClinGenでCurated Pathogenic CNV  
病原性CNVと考えられる

Pathogenicを支持する所見: あり

- Curated Benign: 登録なし
- DGV Gold Standard: 登録なし

Benignを支持する所見: なし



Pathogenic CNV

この動画ではマイクロアレイ染色体検査（以下、CMA）の「結果を見る」「結果を解釈する」ために必要な最低限の基本事項を述べています。

データベースの使用方法は様々なアレンジがあります。実践しながらオリジナルの使用方法を確立していただければと思います。