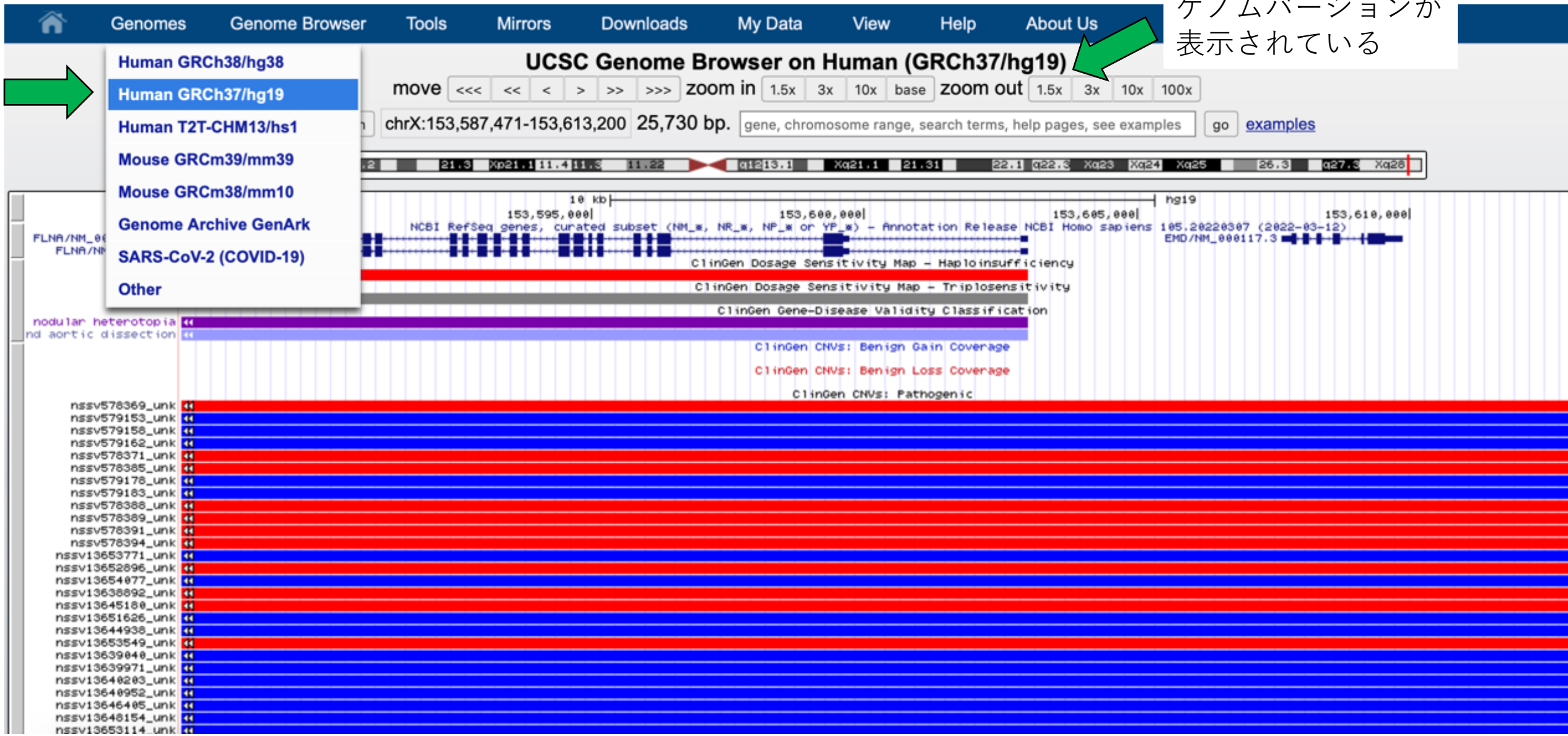
マイクロアレイ染色体検査普及のための産学連携コンソーシアム主催
マイクロアレイ染色体検査解釈ハンズオンウェビナー 2023.9.19

マイクロアレイ染色体検査解釈ハンズオンウェビナー

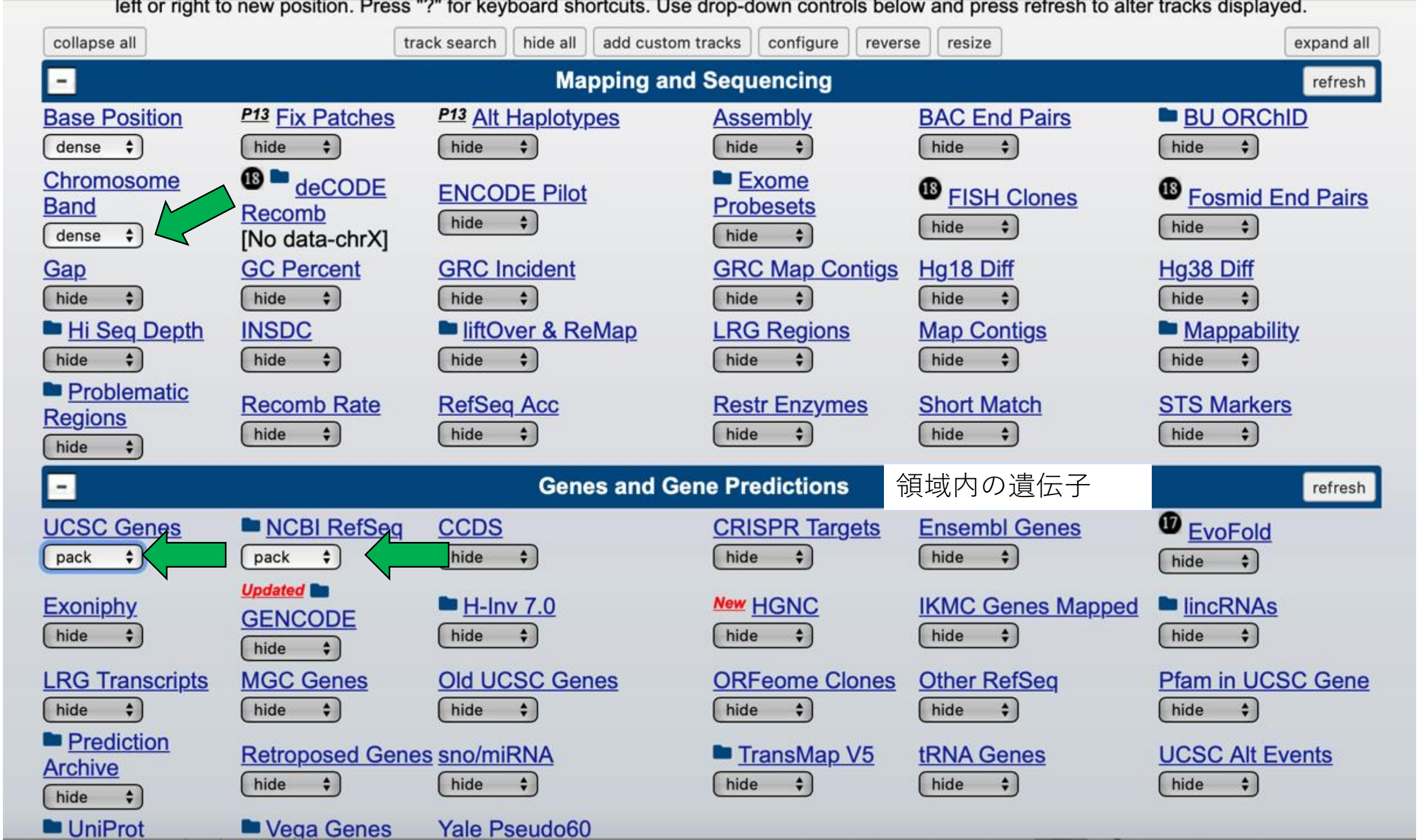
初級編　解説資料

神奈川県立こども医療センター遺伝科
黒田　友紀子

UCSC genome browser の活用



ゲノムバージョンが表示されている


left or right to new position. Press "?" for keyboard shortcuts. Use drop-down controls below and press refresh to alter tracks displayed.
collapse all
track search
hide all
add custom tracks
configure
reverse
resize
expand all
Mapping and Sequencing
refresh
Base Position
dense
P13 Fix Patches
hide
P13 Alt Haplotypes
hide
Assembly
hide
BAC End Pairs
hide
BU ORChID
hide
Chromosome Band
dense
deCODE Recomb
[No data-chrX]
ENCODE Pilot
hide
Exome Probesets
hide
FISH Clones
hide
Fosmid End Pairs
hide
Gap
hide
GC Percent
hide
GRC Incident
hide
GRC Map Contigs
hide
Hg18 Diff
hide
Hg38 Diff
hide
Hi Seq Depth
hide
INSDC
hide
liftOver & ReMap
hide
LRG Regions
hide
Map Contigs
hide
Mappability
hide
Problematic Regions
hide
Recomb Rate
hide
RefSeq Acc
hide
Restr Enzymes
hide
Short Match
hide
STS Markers
hide
Genes and Gene Predictions
refresh
UCSC Genes
pack
NCBI RefSeq
pack
CCDS
hide
CRISPR Targets
hide
Ensembl Genes
hide
EvoFold
hide
Exoniphy
hide
Updated GENCODE
hide
H-Inv 7.0
hide
New HGNC
hide
IKMC Genes Mapped
hide
lincRNAs
hide
LRG Transcripts
hide
MGC Genes
hide
Old UCSC Genes
hide
ORFeome Clones
hide
Other RefSeq
hide
Pfam in UCSC Gene
hide
Prediction Archive
hide
Retroposed Genes
hide
sno/miRNA
hide
TransMap V5
hide
tRNA Genes
hide
UCSC Alt Events
hide
UniProt
Vega Genes
Yale Pseudo60

領域内の遺伝子


Phenotype and Literature
refresh
病原性のあるCNV
Publications hide
CADD hide
ClinGen pack
ClinGen CNVs pack
ClinVar Variants hide
Updated Constraint scores hide
Coriell CNVs hide
COSMIC Regions hide
DECIPHER CNVs pack
DECIPHER SNVs hide
Development Delay pack
GAD View hide
GenCC hide
Gene Interactions hide
GeneReviews hide
GWAS Catalog hide
Haploinsufficiency pack
HGMD Variants hide
Lens Patents hide
LOVD Variants hide
MGI Mouse QTL hide
OMIM Alleles hide
OMIM Cyto Loci hide
OMIM Genes pack
Orphanet hide
New PanelApp hide
REVEL Scores hide
RGD Human QTL hide
RGD Rat QTL hide
SNPedia hide
UniProt Variants hide
Variants in Papers hide
Web Sequences hide
(黄色矢印）クリックすると項目の詳細や細かい設定ができる
COVID-19
refresh
COVID GWAS v4
[No data-chrX]
COVID GWAS v3
[No data-chrX]
Rare Harmful Vars hide
mRNA and EST
refresh
CGAP SAGE hide
Gene Bounds hide
H-Inv hide
Human ESTs hide
Human mRNAs hide
Human RNA Editing hide
Other ESTs
Other mRNAs
Poly(A)
PolyA-Seq
SIB Alt-Splicing
Spliced ESTs

Select subtracks by evidence and class:

All / Class	Evidence: Curated	Evidence: Submitted
Pathogenic	☑	☑
Likely Pathogenic		☑
Uncertain		☑
Likely Benign		☑
Benign	☑	☑

ClinGen CNVs

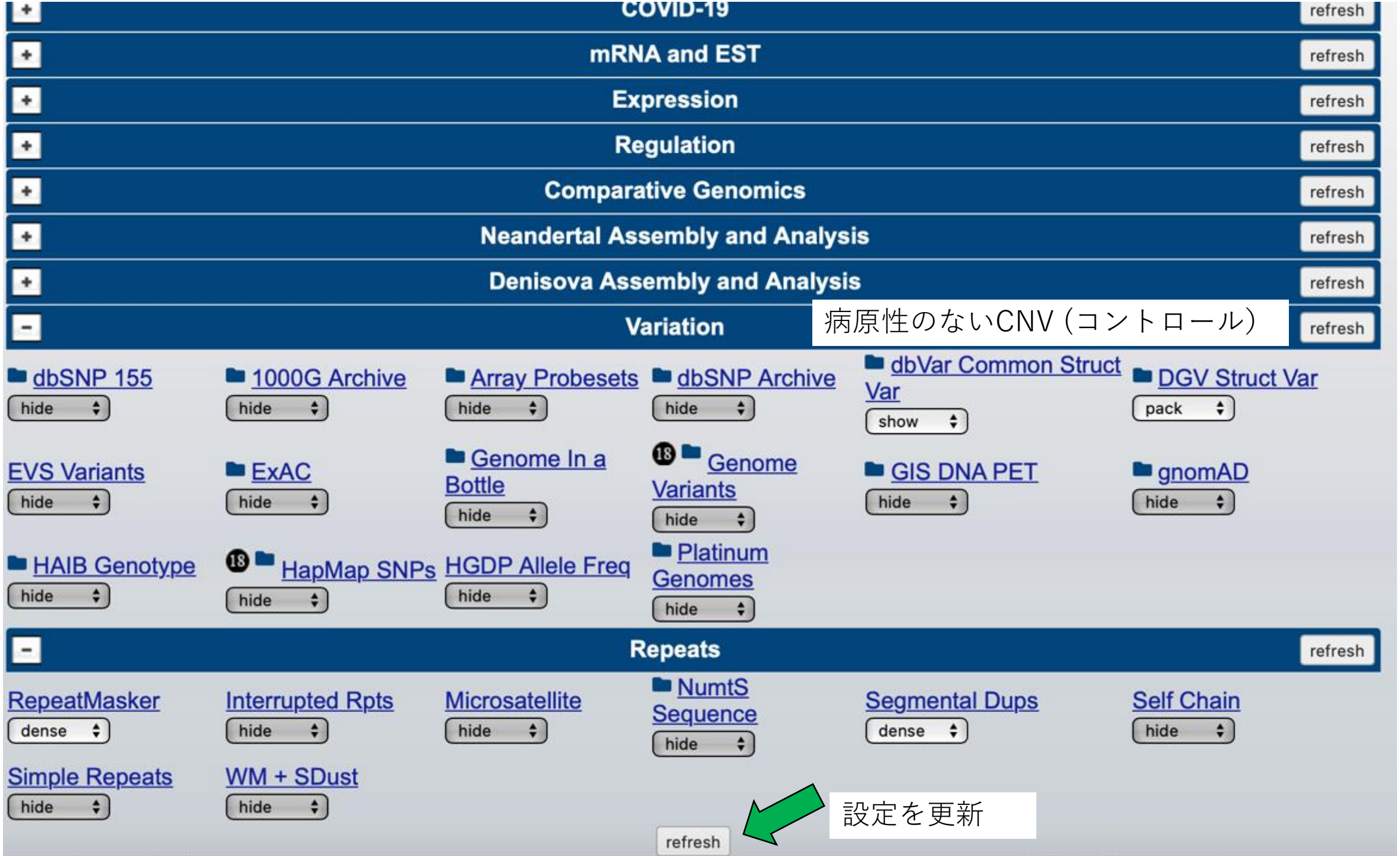
Defaultだとpathogenicしか表示されないようになっているのでbenign, uncertainを表示させたいならpackに変更を

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

		Class↓1	Evidence↓2	views↓3	Track Name↓4	
☑	pack	Benign	Curated	CNVs	ClinGen CNVs: Curated Benign	Schema
☑	pack	Benign	Submitted	CNVs	ClinGen CNVs: Benign	Schema
☑	pack Configure	Benign	Submitted	Coverage	ClinGen CNVs: Benign Gain Coverage	Schema
☑	pack Configure	Benign	Submitted	Coverage	ClinGen CNVs: Benign Loss Coverage	Schema
☑	pack	Likely Benign	Submitted	CNVs	ClinGen CNVs: Uncertain: Likely Benign	Schema
☑	pack	Likely Pathogenic	Submitted	CNVs	ClinGen CNVs: Uncertain: Likely Pathogenic	Schema
☑	pack	Pathogenic	Curated	CNVs	ClinGen CNVs: Curated Pathogenic	Schema
☑	pack	Pathogenic	Submitted	CNVs	ClinGen CNVs: Pathogenic	Schema
☑	pack Configure	Pathogenic	Submitted	Coverage	ClinGen CNVs: Pathogenic Gain Coverage	Schema
☑	pack Configure	Pathogenic	Submitted	Coverage	ClinGen CNVs: Pathogenic Loss Coverage	Schema
☑	pack	Uncertain	Submitted	CNVs	ClinGen CNVs: Uncertain	Schema

11 of 11 selected

Submit


COVID-19
refresh
mRNA and EST
refresh
Expression
refresh
Regulation
refresh
Comparative Genomics
refresh
Neandertal Assembly and Analysis
refresh
Denisova Assembly and Analysis
refresh
Variation
refresh
病原性のないCNV（コントロール）
dbSNP 155
hide
1000G Archive
hide
Array Probesets
hide
dbSNP Archive
hide
dbVar Common Struct Var
show
DGV Struct Var
pack
EVS Variants
hide
ExAC
hide
Genome In a Bottle
hide
Genome Variants
hide
GIS DNA PET
hide
gnomAD
hide
HAIB Genotype
hide
HapMap SNPs
hide
HGDP Allele Freq
hide
Platinum Genomes
hide
Repeats
refresh
RepeatMasker
dense
Interrupted Rpts
hide
Microsatellite
hide
NumtS Sequence
hide
Segmental Dups
dense
Self Chain
hide
Simple Repeats
hide
WM + SDust
hide
refresh
設定を更新