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

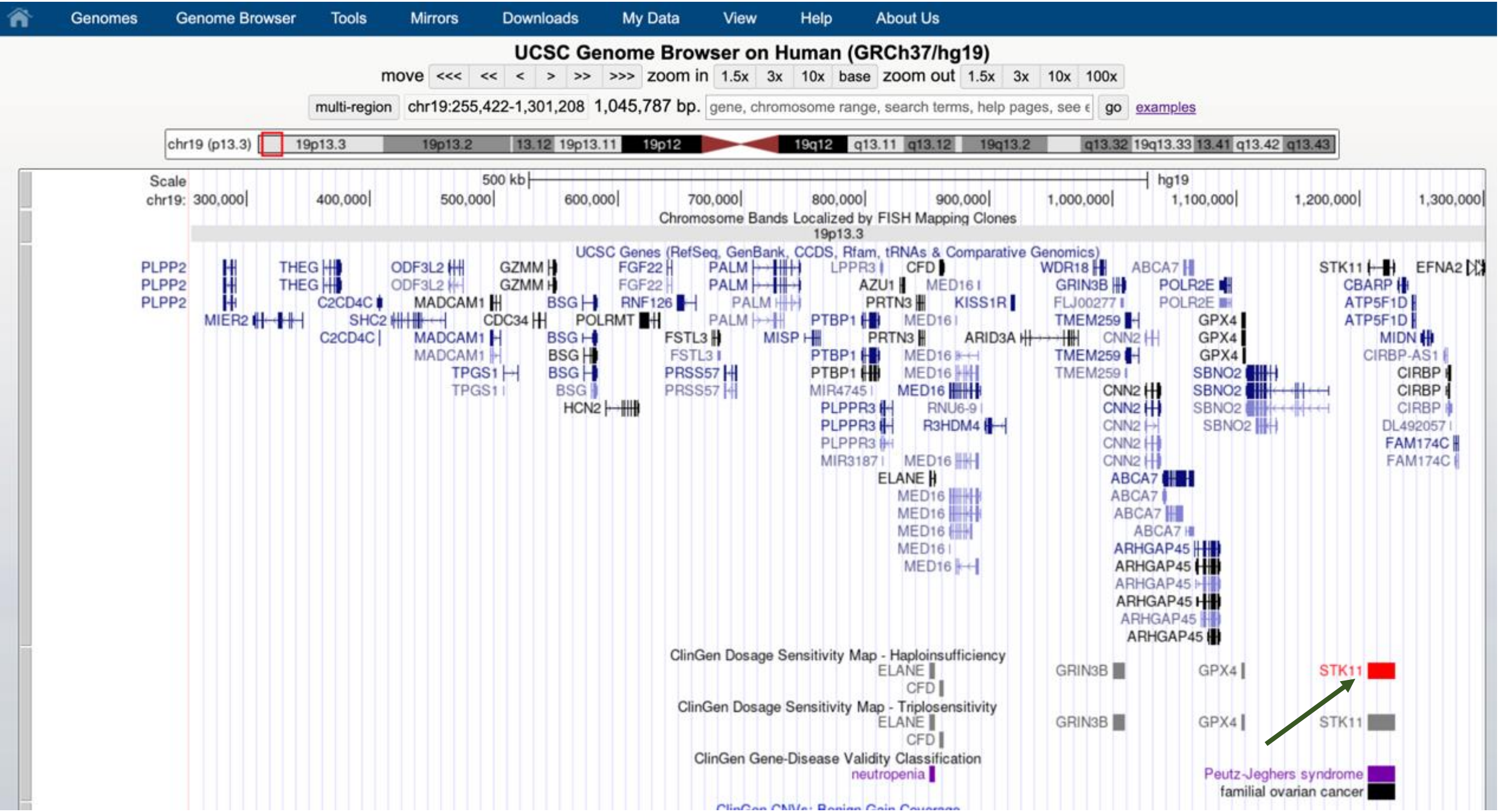
初級編　解説資料 症例３

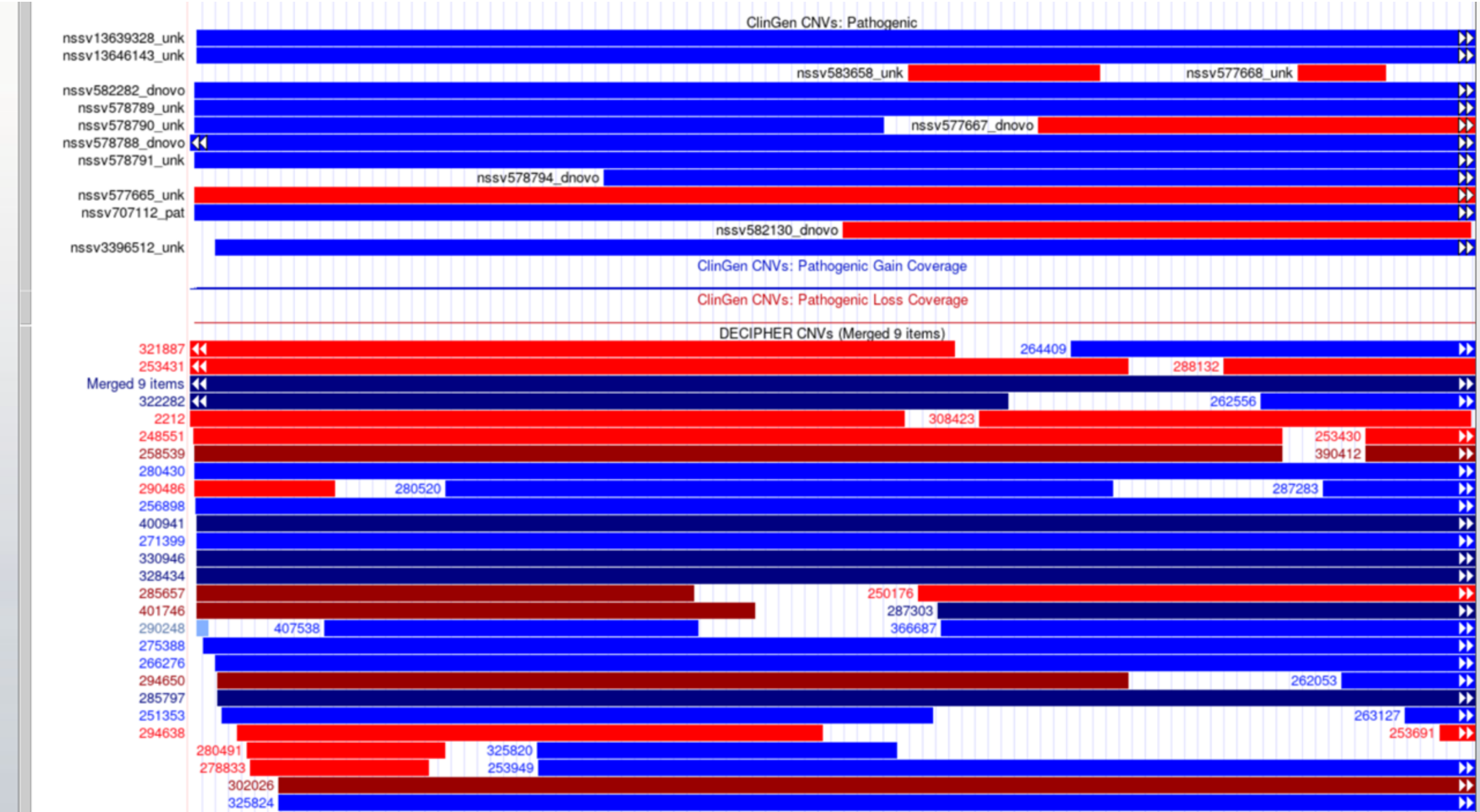
神奈川県立こども医療センター遺伝科
黒田　友紀子

症例3

- 3歳女児：軽度知的障害、斜視
- 【CNV】chr19:255,422-1,301,208
  19p13.3 log2ratio=-0.98 [Loss]
  1.05Mb deletion

- ＊追加設問：遺伝カウンセリングにおける留意点を述べよ


Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
View
Help
About Us
UCSC Genome Browser on Human (GRCh37/hg19)
move <<< << < > >> >>> zoom in 1.5x 3x 10x base zoom out 1.5x 3x 10x 100x
multi-region
chr19:255,422-1,301,208 1,045,787 bp.
gene, chromosome range, search terms, help pages, see e
go
examples
chr19 (p13.3)
19p13.3
19p13.2
13.12
19p13.11
19p12
19q12
q13.11
q13.12
19q13.2
q13.32
19q13.33
13.41
q13.42
q13.43
Scale
chr19:
500 kb
hg19
300,000
400,000
500,000
600,000
700,000
800,000
900,000
1,000,000
1,100,000
1,200,000
1,300,000
Chromosome Bands Localized by FISH Mapping Clones
19p13.3
UCSC Genes (RefSeq, GenBank, CCDS, Rfam, tRNAs & Comparative Genomics)
PLPP2
THEG
ODF3L2
GZMM
FGF22
PALM
LPPR3
CFD
WDR18
ABCA7
STK11
EFNA2
AZU1
MED16
GRIN3B
POLR2E
CBARP
MIER2
SHC2
CDC34
POLRMT
RNF126
BSG
C2CD4C
MADCAM1
PRTN3
KISS1R
FLJ00277
ATP5F1D
PTBP1
TMEM259
GPX4
FSTL3
MISP
ARID3A
CNN2
MIDN
TPGS1
PRSS57
SBNO2
CIRBP-AS1
HCN2
MIR4745
CIRBP
PLPPR3
RNU6-9
R3HDM4
DL492057
FAM174C
MIR3187
ELANE
ARHGAP45
ClinGen Dosage Sensitivity Map - Haploinsufficiency
ClinGen Dosage Sensitivity Map - Triplosensitivity
ClinGen Gene-Disease Validity Classification
neutropenia
Peutz-Jeghers syndrome
familial ovarian cancer

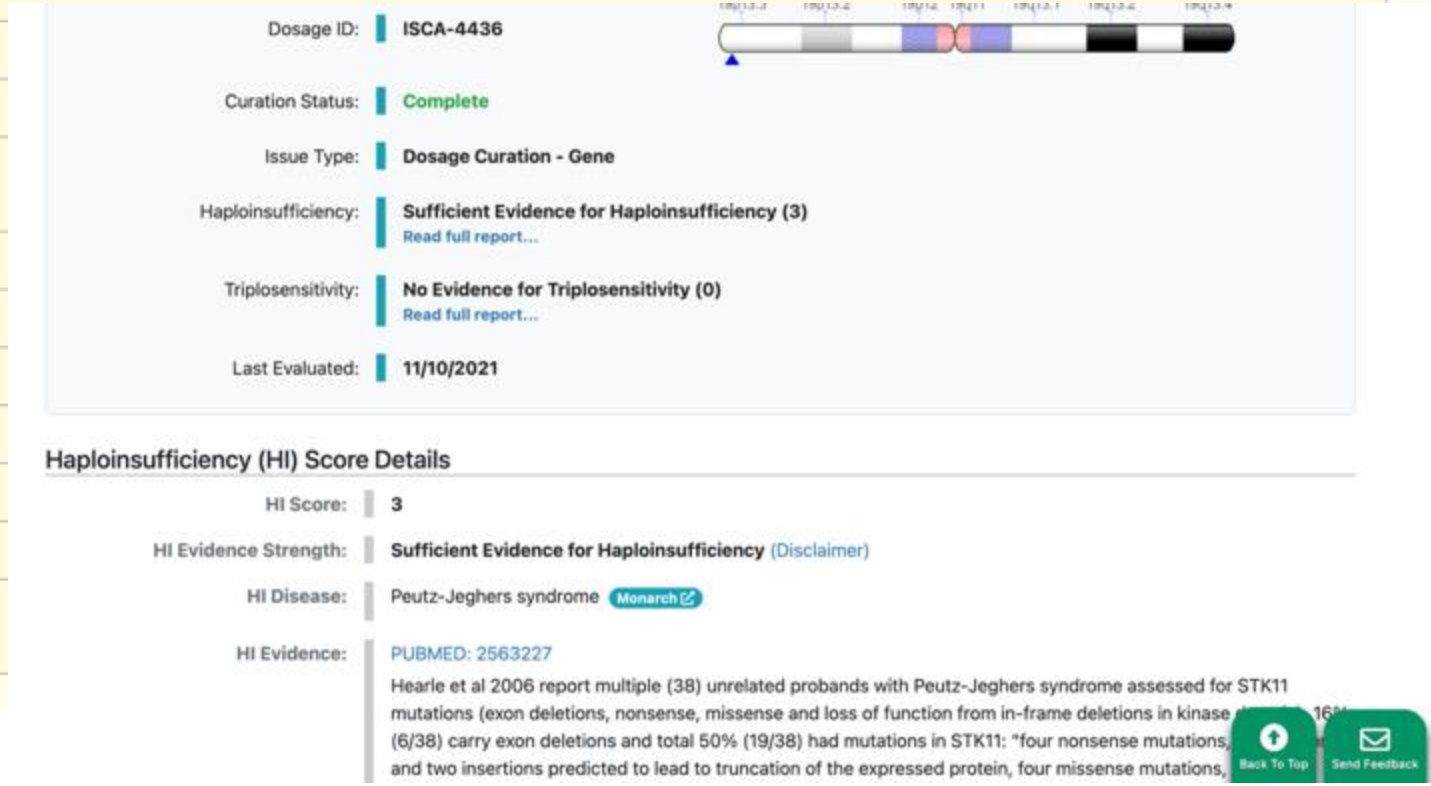

ClinGen CNVs: Pathogenic
nssv13639328_unk
nssv13646143_unk
nssv583658_unk
nssv577668_unk
nssv582282_dnovo
nssv578789_unk
nssv578790_unk
nssv577667_dnovo
nssv578788_dnovo
nssv578791_unk
nssv578794_dnovo
nssv577665_unk
nssv707112_pat
nssv582130_dnovo
nssv3396512_unk
ClinGen CNVs: Pathogenic Gain Coverage
ClinGen CNVs: Pathogenic Loss Coverage
DECIPHER CNVs (Merged 9 items)
321887
264409
253431
288132
Merged 9 items
322282
262556
2212
308423
248551
253430
258539
390412
280430
290486
280520
287283
256898
400941
271399
330946
328434
285657
250176
401746
287303
290248
407538
366687
275388
266276
294650
262053
285797
251353
263127
294638
253691
280491
325820
278833
253949
302026
325824


Haploinsufficiency predictions for genes from DECIPHER
PPAP2C
THEG
ODF3L2
GZMM
FGF22
PALM
AZU1
KISS1R
WDR19
HMHA1
STK11
MUM1
MIER2
SHC2
CDC34
POLRMT
MISP
PRTN3
ARID3A
ABCA7
C19orf26
C2CD4C
MADCAM1
BSG
RNF126
PTBP1
CFD
GRIN3B
POLR2E
ATP5D
TPGS1
HCN2
FSTL3
ELANE
TMEM259
GPX4
MIDN
PRSS57
MED16
CNN2
SBNO2
CIRBP
R3HDM4
C19orf24
EFNA2
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
607126
609503
102670
109480
615177
615289
162815
604161
620291
601155
602216
602756
620092
610336
116948
601778
608134
610391
603265
605414
603150
605217
600311
605831
600693
606651
180664
606700
602781
605343
177020
611011
138322
602649
130130
602373
615729
134350
604062
Database of Genomic Variants: Structural Var Regions (CNV, Inversion, In/del)
nsv471520
nsv9659
esv7704
esv28250
nsv954667
esv2717767
nsv4263752
nsv2385
nsv138039
esv26912
nsv480760
nsv953941
dgv3419n100
esv8994
esv4763
dgv478e201
esv3037
nsv960769
nsv828353
nsv9662
esv2763195
nsv577980
esv28823
nsv828349
esv27631
nsv1121960
nsv954655
esv998260
esv3453
esv25790
nsv516646
nsv828368
nsv962991
nsv1077856
esv26032
dgv6166n54
nsv3362271
nsv3368643
nsv4263394
nsv1072293
esv2717812
esv2586842
nsv476585
esv1002673
esv990677
esv2758742
esv23197
esv21617
nsv482205
nsv1072832
nsv828354
esv2665746
nsv4576702
nsv828367
nsv512557
nsv138017
nsv428360
nsv3376055
nsv4262929
nsv4570805
nsv578190
nsv475858
nsv578268
nsv833702
esv33417
esv3656019
nsv4255387
nsv4265958
esv2717800
nsv3368325
nsv960770
nsv2386
esv9337
nsv960886
nsv1071479
esv3494
esv2717714
nsv828350
esv2717768
nsv4272442
esv3656792
nsv4258754
nsv828360
nsv953939
esv24381
esv3643341
nsv9660
nsv2384
nsv138339
dgv134n111
nsv470102
nsv578272
esv2422333
esv3574264
nsv4253805
nsv4261111
nsv4531700
nsv3359646
nsv3368810
nsv512556
nsv578216
dgv6196n54
nsv4531408
esv26691
nsv3365632
esv26031
nsv578164
esv3643369
nsv954613
nsv482206
nsv4531317
nsv470103
nsv1058440
esv27916
nsv3378131
nsv3377821
nsv578163
nsv4261690
esv3656119
nsv4257081
esv2044506
esv3656816
esv2717833
nsv4268781
nsv960887
nsv4262887
esv2323487
nsv3361746
nsv828351
nsv3362806
nsv458321
esv3643377
nsv4268520
dgv6185n54
nsv1071886
nsv3358271
nsv1134736
nsv828348
nsv3360547
nsv1116526
nsv4257547
esv2012083
esv3331953
nsv469845
nsv1060536
nsv4506158
nsv1150208
esv5410
nsv3371524
esv3417596
esv3670093
esv3659301
nsv3377722
esv2717811
nsv3351488
dgv6186n54
nsv1143897
esv3643342
nsv578092
nsv4258802
nsv3361235
nsv3360130
nsv4271635
nsv3359222
esv1501734
esv22453
nsv526080
nsv4258123
nsv9652
nsv509711
nsv477965
esv2717732
nsv4270482
esv1111384
nsv578167
esv3659305
esv3670096
esv2463047
nsv4263050
esv25497
nsv578123
nsv3359135
nsv4271033
nsv4265380
esv3473
nsv83370
esv2060795
nsv953938
esv3643390
nsv138494
nsv4265927
nsv138298
esv2717724
esv2717756
dgv1677n106
dgv566n67
nsv1112732
nsv4256657
nsv3354450
esv2135609
nsv4262630
esv2717721
nsv4515003
esv3643362
nsv4532385
esv28908
esv2717835
nsv1072827
nsv3370427
nsv3365882
nsv3362991
nsv4256928
nsv3364441
nsv578168
nsv4267956
nsv1143893
nsv4263175
nsv138169
nsv828369
nsv1123202
nsv138274
esv3643351
esv2717728
esv3643357
esv2717781
nsv3375160
esv3656185
nsv3350596
esv3656828
esv2717836
esv1162378
esv3555808
nsv3369515
nsv3368390
esv3643358
esv2717782
nsv138320
nsv828358
nsv578189
nsv828366
esv2717832
esv3643391
nsv1125391
nsv3366829
nsv4531139
esv1634833
nsv4531620
nsv458320
nsv4255706
nsv4264538
nsv4267468
nsv578215
nsv4269006
esv3656285
nsv3339892
nsv3370150
nsv4258672
nsv3365530
esv2176059
esv3893155
nsv1160555
nsv3357983
esv24777
nsv1123205
nsv3359212
esv2717757
nsv4264207
esv26343
esv3659304
esv3643383
nsv513506
esv2717837
ID:nsv833704; Position;
chr19:951640-1144485; Size:192845;
Type:gain; Affected genes:ABCA7, ARID3A,
CNN2, GPX4, GRIN3B, HMHA1, POLR2E,
SBNO2, TMEM259, WDR18
Database of Genomic Variants: Structural Variation (CNV, Inversion, In/del)
DGV Supp Var
Database of Genomic Variants: Gold Standard Variants
gssvG18146
gssvG18150
gssvG18156
gssvL55557
gssvL55558
gssvL55559
gssvL55560
gssvL55631
gssvL55626
gssvL55578
gssvL55642
gssvL55676
gssvL55846
gssvG18196
gssvG18206
gssvL55572
gssvL55570
gssvL55649
gssvG18193
gssvL55703
gssvL55669
gssvG18176
gssvL55785
gssvG18188
gssvL55667
gssvL55609
gssvL55723
gssvG18194
gssvL55711
gssvL55683
gssvG18184
gssvL55722
gssvL55693
gssvG18212
gssvL55640
gssvL55691
gssvL55610
gssvL55697
gssvL55629
gssvG18154
gssvG18213
gssvL55627
gssvL55661
gssvL55780
gssvL55741
gssvL55851
gssvL55729
gssvL55811
gssvL55735
gssvL55696
gssvG18258
gssvG18231
gssvL55690
gssvL55621
gssvL55816
gssvG18229
gssvG18223
gssvL55634
gssvL55686
gssvL55646
gssvL55738
gssvL55736
gssvG18259
gssvL55632
gssvL55695
gssvG18262
gssvG18251
gssvL55732
gssvL55847
gssvL55656
gssvL55761
Container of all new and previous GENCODE releases - this is a container track with 20 subtracks of different types (super track)
Repeating Elements by RepeatMasker
RepeatMasker
Duplications of >1000 Bases of Non-RepeatMasked Sequence

ClinGen Dosage Sensitivity Map - Haploinsufficiency (STK11)

Item: STK11
Score: 0
Position: chr19:1205798-1228434
Band: 19p13.3
Genomic Size: 22637
View DNA for this feature (hg19/Human)

ClinGen URL	https://search.clinicalgenome.org/kb/gene-dosage/STK11
Gene Symbol or ISCA Region Name	STK11
Gene ID or ISCA ID	6794
Cytoband	19p13.3
Last evaluation date	2021-11-10
Haploinsufficiency score	3
Haploinsufficiency phenotype description	Sufficient evidence for dosage pathogenicity
Associated PubMed ID 1	2563227
Associated PubMed ID 2	20623358
Associated PubMed ID 3	23892522
Associated PubMed ID 4	28231849
Associated PubMed ID 5	
Associated PubMed ID 6	
Mondo disease ontology ID	MONDO:0008280



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19p13.3 deletion STK11

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18 results

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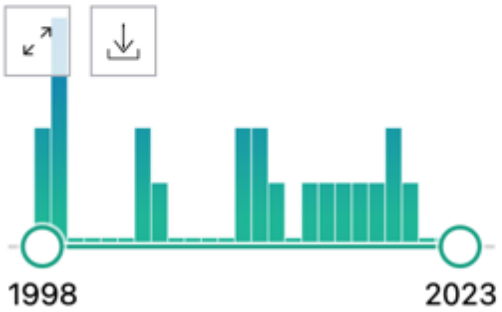
RESULTS BY YEAR



TEXT AVAILABILITY

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- Associated data

1 Cite Share

Chromosome **19p13.3 deletion** in a child with Peutz-Jeghers syndrome, congenital heart defect, high myopia, learning difficulties and dysmorphic features: Clinical and molecular characterization of a new contiguous **gene** syndrome.

Souza J, Faucz F, Sotomaior V, Filho AB, Rosenfeld J, Raskin S.

Genet Mol Biol. 2011 Oct;34(4):557-61. doi: 10.1590/S1415-47572011005000044. Epub 2011 Oct 1.

PMID: 22215957 **Free PMC article.**

This is a report on a girl with PJS features, learning difficulties, dysmorphic features and cardiac malformation, bearing a de novo 1.1 Mb **deletion** at **19p13.3**. This **deletion** encompasses at least 47 genes, including **STK11**. This is the first repo ...

2 Cite Share

Distinct phenotype associated with a cryptic subtelomeric **deletion** of **19p13.3**-pter.

Archer HL, Gupta S, Enoch S, Thompson P, Rowbottom A, Chua I, Warren S, Johnson D, Ledbetter DH, Lese-Martin C, Williams P, Pilz DT.

Am J Med Genet A. 2005 Jul 1;136(1):38-44. doi: 10.1002/ajmg.a.30774.

PMID: 15937949 Review.

回答	症例3
CNV病原性(5段階評価)	Pathogenic
遺伝学的診断	19p13.3微細欠失(Likely Pathogenic) 欠失型Peutz-Jeghers症候群 (Pathogenic)
判断根拠 -使用したデータベースの結果や文献情報等より病原性や遺伝学的診断の判断　根拠を明確に（自由記載）	*STK11*遺伝子以外にClinGen HI遺伝子を含まないが、同領域の欠失で軽度の知的障害を認めた例が複数存在する。 ハプロ不全遺伝子*STK11*を含む欠失であり、欠失型PJS ClinGen Haploinsufficiency score 3 Sufficient Evidence for Haploinsufficiency (3) 35遺伝子以上を含む欠失、複数の文献で知的障害の報告あり
追加質問 ＊両親の表現型はない。推測される遺伝性について述べよ	学童期以降に口唇粘膜疹の出現、消化管の腫瘍サーベイランスの適応である。