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

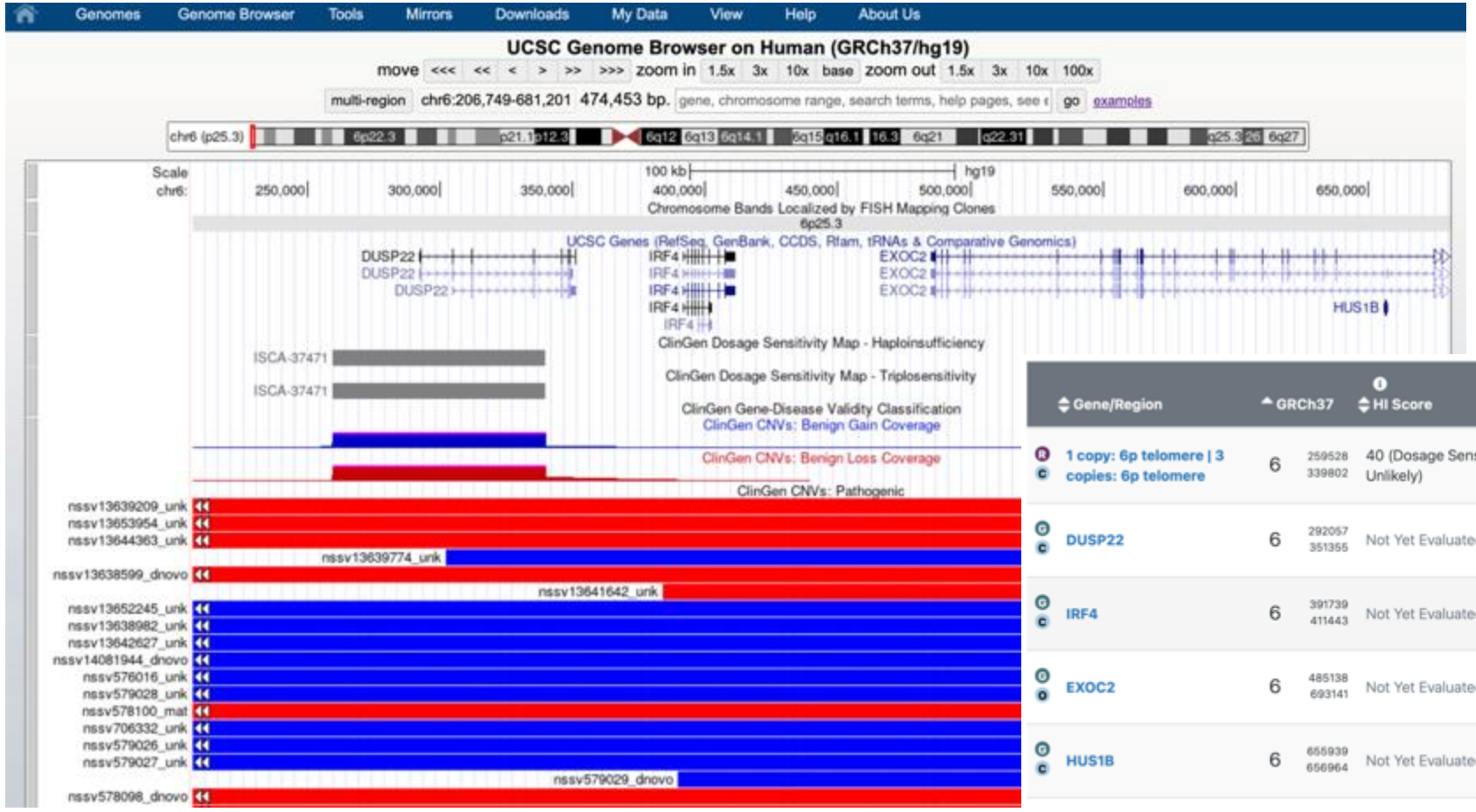
初級編　解説資料 症例４

神奈川県立こども医療センター遺伝科
黒田　友紀子

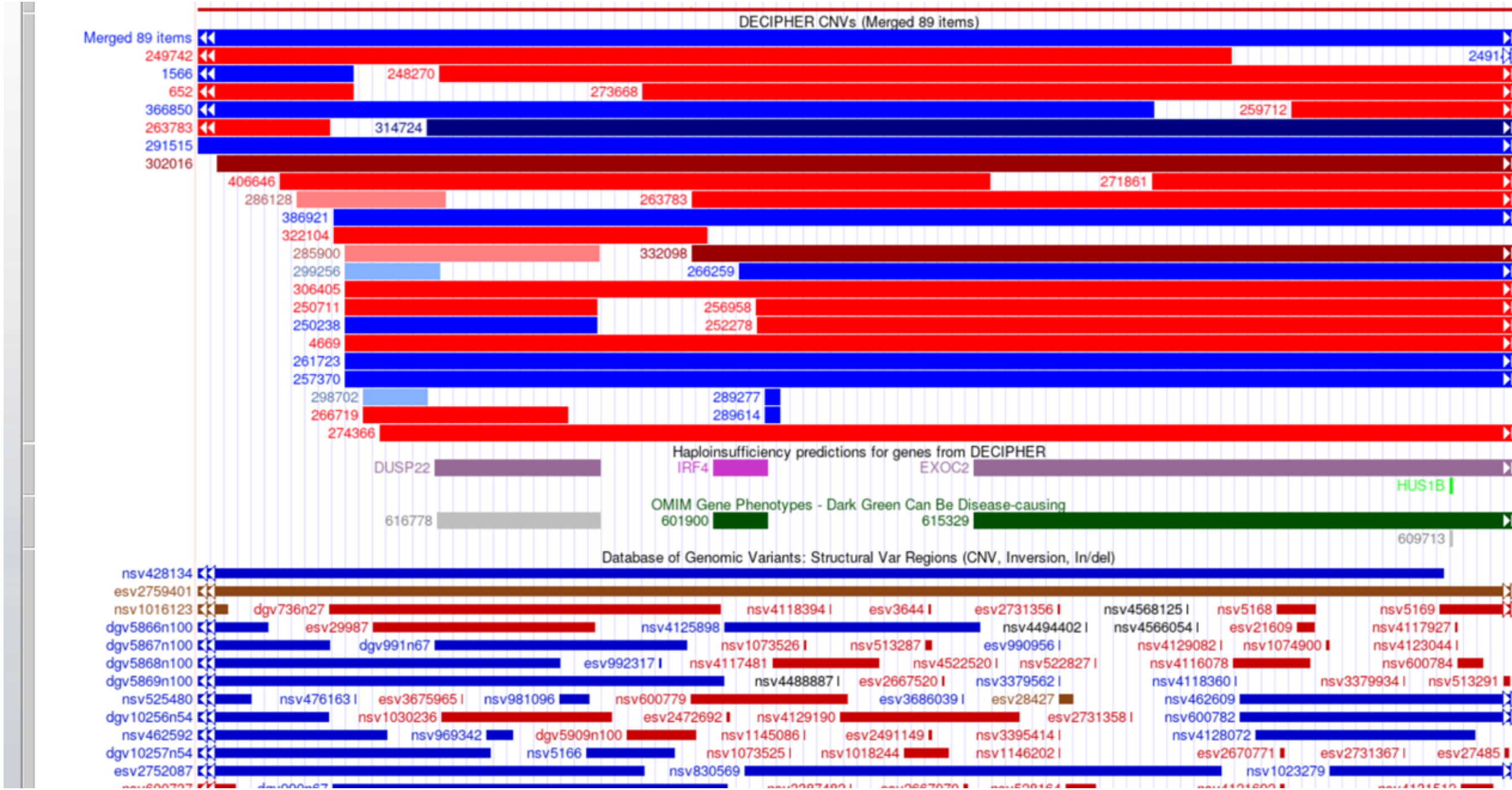
症例4

- 9歳男児：知的障害、鎖肛
- 【CNV1】chr6:206,749-681,201 log2ratio=0.57 [Gain]
- 【CNV2】chr7:155054023-158909738 log2ratio=-1.0 [Loss]
- ＊追加質問：追加で必要な遺伝学的検査について述べよ

【CNV1】chr6:206,749-681,201
log2ratio=0.57 [Gain]
6p25.3-pter 474 kb duplication

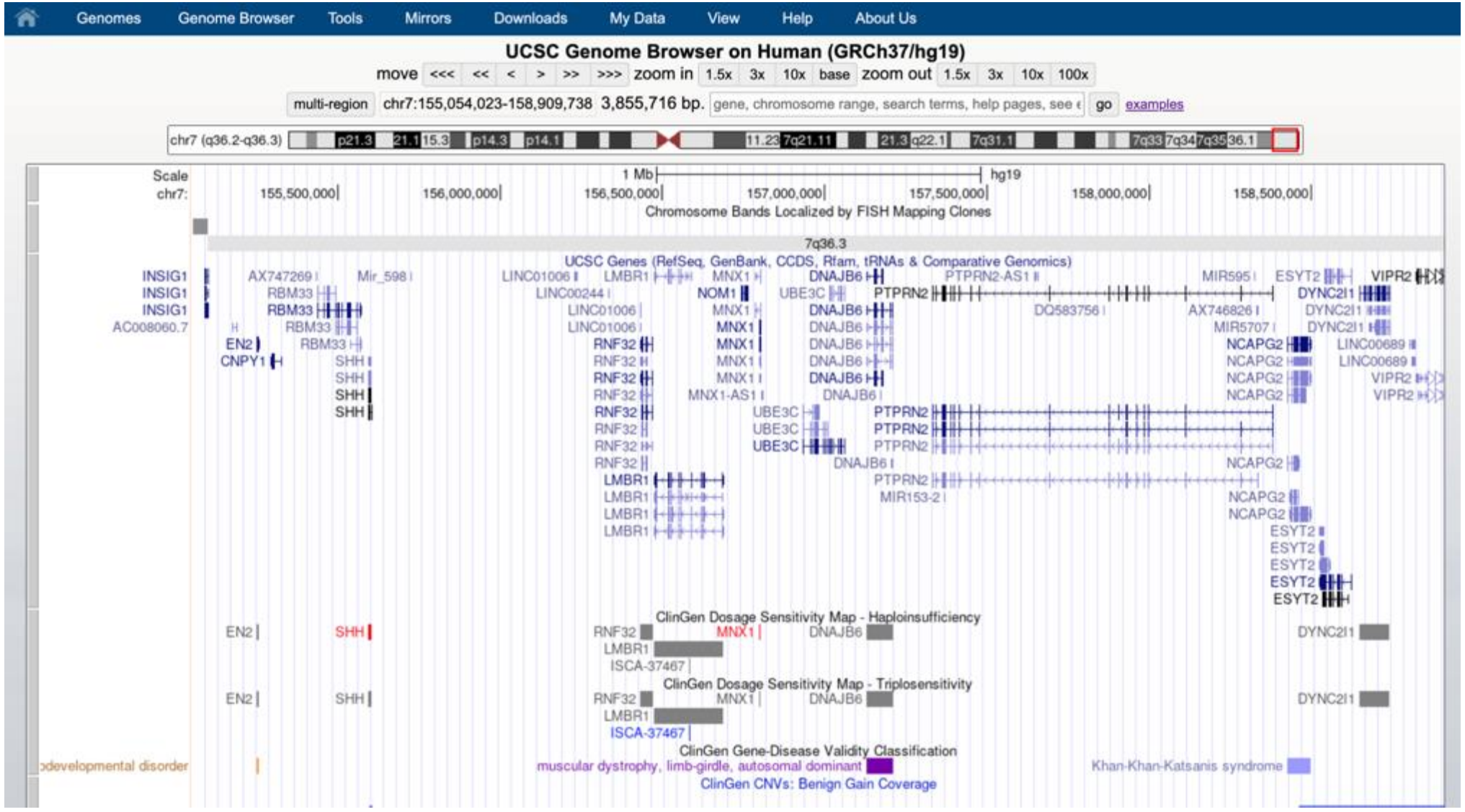


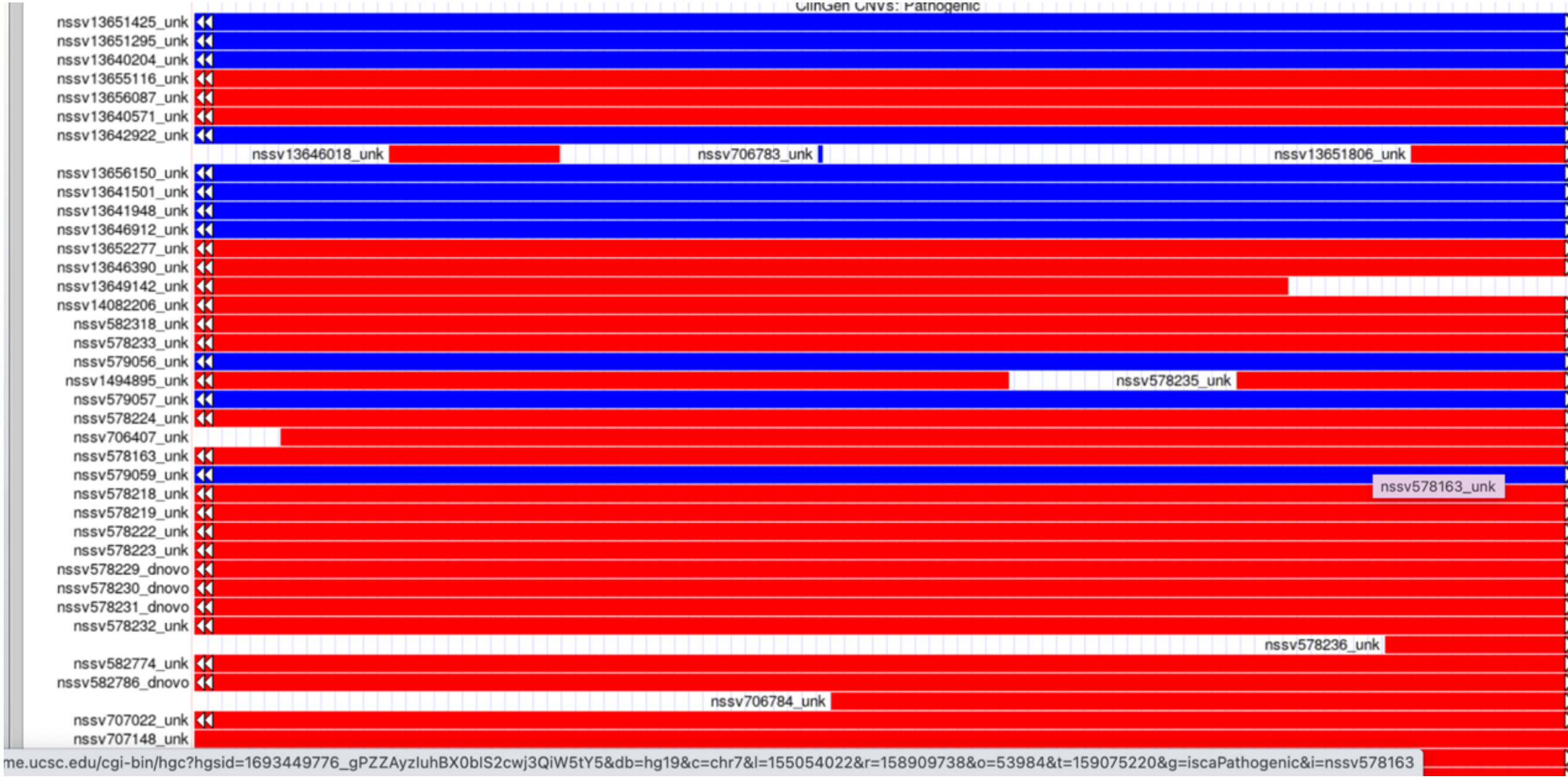
Gene/Region	GRCh37		HI Score	TS Score	OMIM	Morbid	%HI	pLI	LOEUF	Report
1 copy: 6p telomere | 3 copies: 6p telomere	6	259528 339802	40 (Dosage Sensitivity Unlikely)	40 (Dosage Sensitivity Unlikely)			-	-	-	Complete
DUSP22	6	292057 351355	Not Yet Evaluated	Not Yet Evaluated	✓		38.68	0.01	0.93	Awaiting Review
IRF4	6	391739 411443	Not Yet Evaluated	Not Yet Evaluated	✓	✓	19.27	0.86	0.38	Awaiting Review
EXOC2	6	485138 693141	Not Yet Evaluated	Not Yet Evaluated	✓	✓	34.26	0	0.59	Awaiting Review
HUS1B	6	655939 656964	Not Yet Evaluated	Not Yet Evaluated	✓		97.37	0	0	Awaiting Review
LOC100421511	6	669080 670307	-1 (Pseudogene)	-1 (Pseudogene)			-	-	-	Not Reviewable

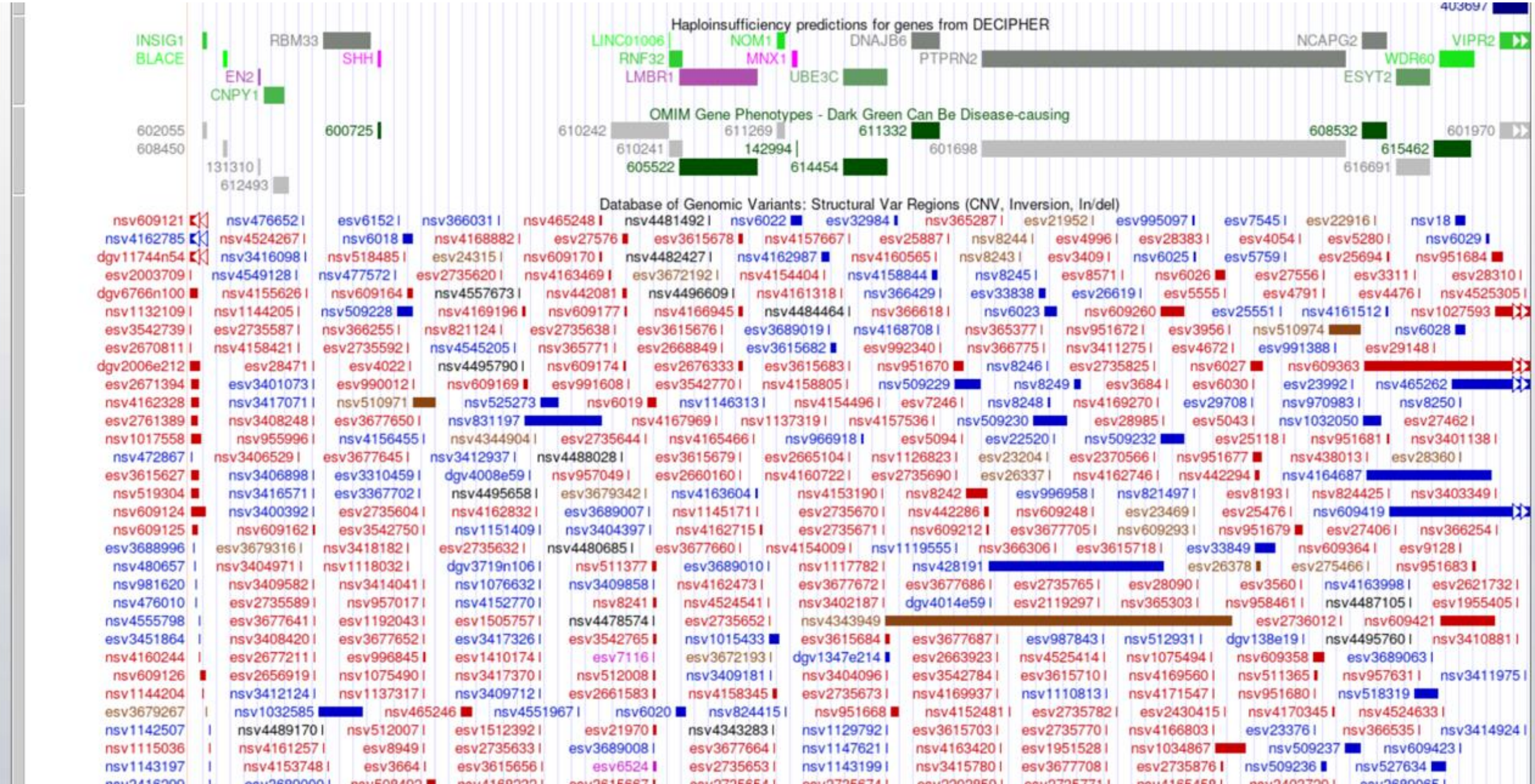

DECIPHER CNVs (Merged 89 items)
Merged 89 items
249742
1566
652
366850
263783
291515
302016
406646
286128
386921
322104
285900
299256
306405
250711
250238
4669
261723
257370
298702
266719
274366
248270
273668
314724
271861
263783
332098
266259
256958
252278
289277
289614
2491
259712
Haploinsufficiency predictions for genes from DECIPHER
DUSP22
IRF4
EXOC2
HUS1B
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
616778
601900
615329
609713
Database of Genomic Variants: Structural Var Regions (CNV, Inversion, In/del)
nsv428134
esv2759401
nsv1016123
dgv5866n100
dgv5867n100
dgv5868n100
dgv5869n100
nsv525480
dgv10256n54
nsv462592
dgv10257n54
esv2752087
dgv736n27
esv29987
dgv991n67
esv992317
nsv476163
esv3675965
nsv981096
nsv1030236
dgv5909n100
nsv969342
nsv5166
nsv830569
nsv4118394
nsv4125898
nsv1073526
nsv4117481
nsv4488887
esv3644
esv2731356
nsv4568125
nsv5168
nsv5169
nsv4494402
nsv4566054
esv21609
nsv4117927
nsv513287
esv990956
nsv4129082
nsv1074900
nsv4123044
nsv4522520
nsv522827
nsv4116078
nsv600784
esv2667520
nsv3379562
nsv4118360
nsv3379934
nsv513291
esv3686039
esv28427
nsv462609
esv2472692
nsv4129190
esv2731358
nsv600782
nsv1145086
esv2491149
nsv3395414
nsv4128072
nsv1073525
nsv1018244
nsv1146202
esv2670771
esv2731367
esv27485
nsv1023279

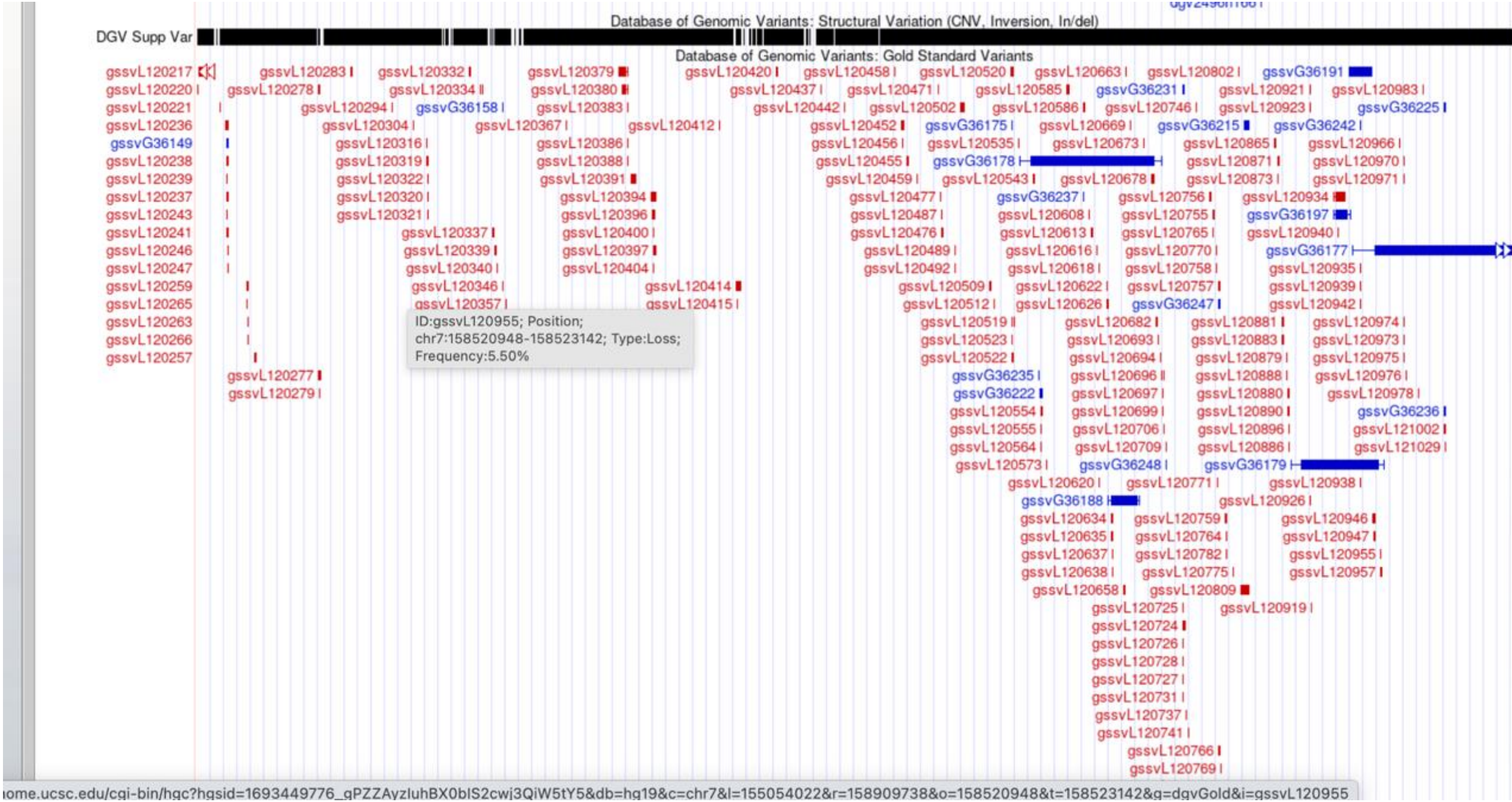

dgv5901n100
nsv1111687
nsv1153784
nsv3389871
esv3567124
nsv1121338
nsv4114982
nsv3386875
Database of Genomic Variants: Structural Variation (CNV, Inversion, In/del)
DGV Supp Var
Database of Genomic Variants: Gold Standard Variants
gssvL104465
gssvG31584
gssvG31576
gssvL104536
gssvL104499
gssvL104501
gssvL104505
gssvL104498
gssvG31563
gssvG31569
gssvL104504
gssvL104503
gssvL104506
gssvL104480
gssvG31571
gssvL104500
gssvG31568
gssvL104469
gssvL104523
gssvG31566
gssvL104519
gssvG31562
gssvL104485
gssvG31565
gssvL104471
gssvL104463
gssvG31561
gssvL104459
gssvL104476
gssvL104464
gssvG31570
Repeating Elements by RepeatMasker
RepeatMasker
Duplications of >1000 Bases of Non-RepeatMasked Sequence
Segmental Dups
Click to alter the display density of Segmental Dups

【CNV2】 chr7:155054023-158909738 7q36.2-q36.3(qter) 3.86Mb deletion

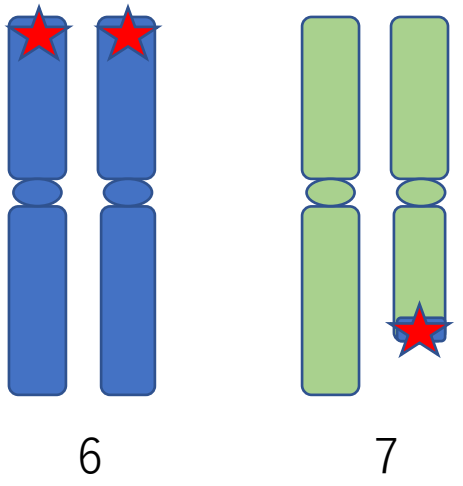



ClinGen CNVs: Pathogenic
nssv13651425_unk
nssv13651295_unk
nssv13640204_unk
nssv13655116_unk
nssv13656087_unk
nssv13640571_unk
nssv13642922_unk
nssv13646018_unk
nssv706783_unk
nssv13651806_unk
nssv13656150_unk
nssv13641501_unk
nssv13641948_unk
nssv13646912_unk
nssv13652277_unk
nssv13646390_unk
nssv13649142_unk
nssv14082206_unk
nssv582318_unk
nssv578233_unk
nssv579056_unk
nssv1494895_unk
nssv578235_unk
nssv579057_unk
nssv578224_unk
nssv706407_unk
nssv578163_unk
nssv579059_unk
nssv578218_unk
nssv578163_unk
nssv578219_unk
nssv578222_unk
nssv578223_unk
nssv578229_dnovo
nssv578230_dnovo
nssv578231_dnovo
nssv578232_unk
nssv578236_unk
nssv582774_unk
nssv582786_dnovo
nssv706784_unk
nssv707022_unk
nssv707148_unk
me.ucsc.edu/cgi-bin/hgc?hgsid=1693449776_gPZZAyzIuhBX0bIS2cwj3QiW5tY5&db=hg19&c=chr7&l=155054022&r=158909738&o=53984&t=159075220&g=iscaPathogenic&i=nssv578163

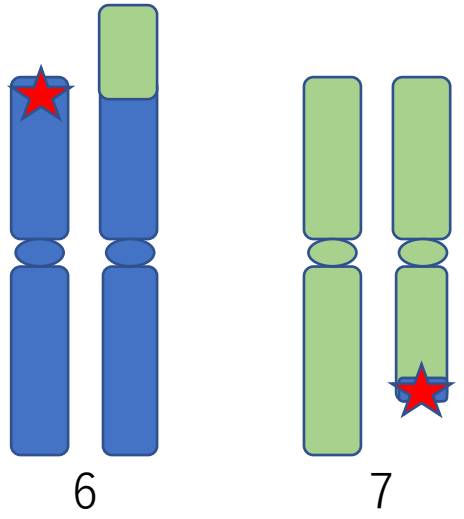

Haploinsufficiency predictions for genes from DECIPHER
INSIG1
BLACE
RBM33
SHH
EN2
CNPY1
LINC01006
RNF32
NOM1
MNX1
LMBR1
DNAJB6
UBE3C
NCAPG2
PTPRN2
VIPR2
WDR60
ESYT2
OMIM Gene Phenotypes - Dark Green Can Be Disease-causing
602055
608450
131310
612493
600725
610242
610241
611269
142994
611332
605522
614454
608532
601698
601970
615462
616691
Database of Genomic Variants: Structural Var Regions (CNV, Inversion, In/del)


Database of Genomic Variants: Structural Variation (CNV, Inversion, In/del)
DGV Supp Var
Database of Genomic Variants: Gold Standard Variants
gssvL120217
gssvL120283
gssvL120332
gssvL120379
gssvL120420
gssvL120458
gssvL120520
gssvL120663
gssvL120802
gssvG36191
gssvL120220
gssvL120278
gssvL120334
gssvL120380
gssvL120437
gssvL120471
gssvL120585
gssvG36231
gssvL120921
gssvL120983
gssvL120221
gssvL120294
gssvG36158
gssvL120383
gssvL120442
gssvL120502
gssvL120586
gssvL120746
gssvL120923
gssvG36225
gssvL120236
gssvL120304
gssvL120367
gssvL120412
gssvL120452
gssvG36175
gssvL120669
gssvG36215
gssvG36242
gssvG36149
gssvL120316
gssvL120386
gssvL120456
gssvL120535
gssvL120673
gssvL120865
gssvL120966
gssvL120238
gssvL120319
gssvL120388
gssvL120455
gssvG36178
gssvL120871
gssvL120970
gssvL120239
gssvL120322
gssvL120391
gssvL120459
gssvL120543
gssvL120678
gssvL120873
gssvL120971
gssvL120237
gssvL120320
gssvL120394
gssvL120477
gssvG36237
gssvL120756
gssvL120934
gssvL120243
gssvL120321
gssvL120396
gssvL120487
gssvL120608
gssvL120755
gssvG36197
gssvL120241
gssvL120337
gssvL120400
gssvL120476
gssvL120613
gssvL120765
gssvL120940
gssvL120246
gssvL120339
gssvL120397
gssvL120489
gssvL120616
gssvL120770
gssvG36177
gssvL120247
gssvL120340
gssvL120404
gssvL120492
gssvL120618
gssvL120758
gssvL120935
gssvL120259
gssvL120346
gssvL120414
gssvL120509
gssvL120622
gssvL120757
gssvL120939
gssvL120265
gssvL120357
gssvL120415
gssvL120512
gssvL120626
gssvG36247
gssvL120942
gssvL120263
gssvL120519
gssvL120682
gssvL120881
gssvL120974
gssvL120266
gssvL120523
gssvL120693
gssvL120883
gssvL120973
gssvL120257
gssvL120522
gssvL120694
gssvL120879
gssvL120975
gssvL120277
gssvG36235
gssvL120696
gssvL120888
gssvL120976
gssvL120279
gssvG36222
gssvL120697
gssvL120880
gssvL120978
gssvL120554
gssvL120699
gssvL120890
gssvG36236
gssvL120555
gssvL120706
gssvL120896
gssvL121002
gssvL120564
gssvL120709
gssvL120886
gssvL121029
gssvL120573
gssvG36248
gssvG36179
gssvL120620
gssvL120771
gssvL120938
gssvG36188
gssvL120926
gssvL120634
gssvL120759
gssvL120946
gssvL120635
gssvL120764
gssvL120947
gssvL120637
gssvL120782
gssvL120955
gssvL120638
gssvL120775
gssvL120957
gssvL120658
gssvL120809
gssvL120725
gssvL120919
gssvL120724
gssvL120726
gssvL120728
gssvL120727
gssvL120731
gssvL120737
gssvL120741
gssvL120766
gssvL120769
ID:gssvL120955; Position;
chr7:158520948-158523142; Type:Loss;
Frequency:5.50%
ome.ucsc.edu/cgi-bin/hgc?hgsid=1693449776_qPZZAyzIuhBX0bIS2cwj3QiW5tY5&db=hg19&c=chr7&l=155054022&r=158909738&o=158520948&t=158523142&g=dgvGold&i=gssvL120955

回答	CNV1 chr6:206,749-681,201 6p25.3-pter 474 kb duplication	CNV2 chr7:155,054,023-158,909,738 7q36.2-q36.3(qter)　3.86Mb deletion
CNV病原性	Likely benign	Pathogenic
遺伝学的診断	6番染色体短腕部分トリソミー	7番染色体長腕端部部分モノソミー
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）	*DUSP22,IRF4, EXOC2* 遺伝子のみを含む重複、明らかなTriplosensitivity geneを含まない。DGVでの登録あり。 Pathogenicの登録はない。	HI遺伝子である*SHH, MNX1*遺伝子(HI score 3) が欠失している。 3Mb以上のサイズの欠失でOMIM geneを17個含み、明らかなbenign CNVとして登録がない。
追加質問	追加で必要な遺伝学的検査: G-band染色体検査、7q(+6p)サブテロメアFISH 予想される核型: 46,XY,der(7)t(6;7)(p25.3;q36.2)	



予想される核型



両親のいずれかが
均衡型相互転座保因者の
可能性がある