

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

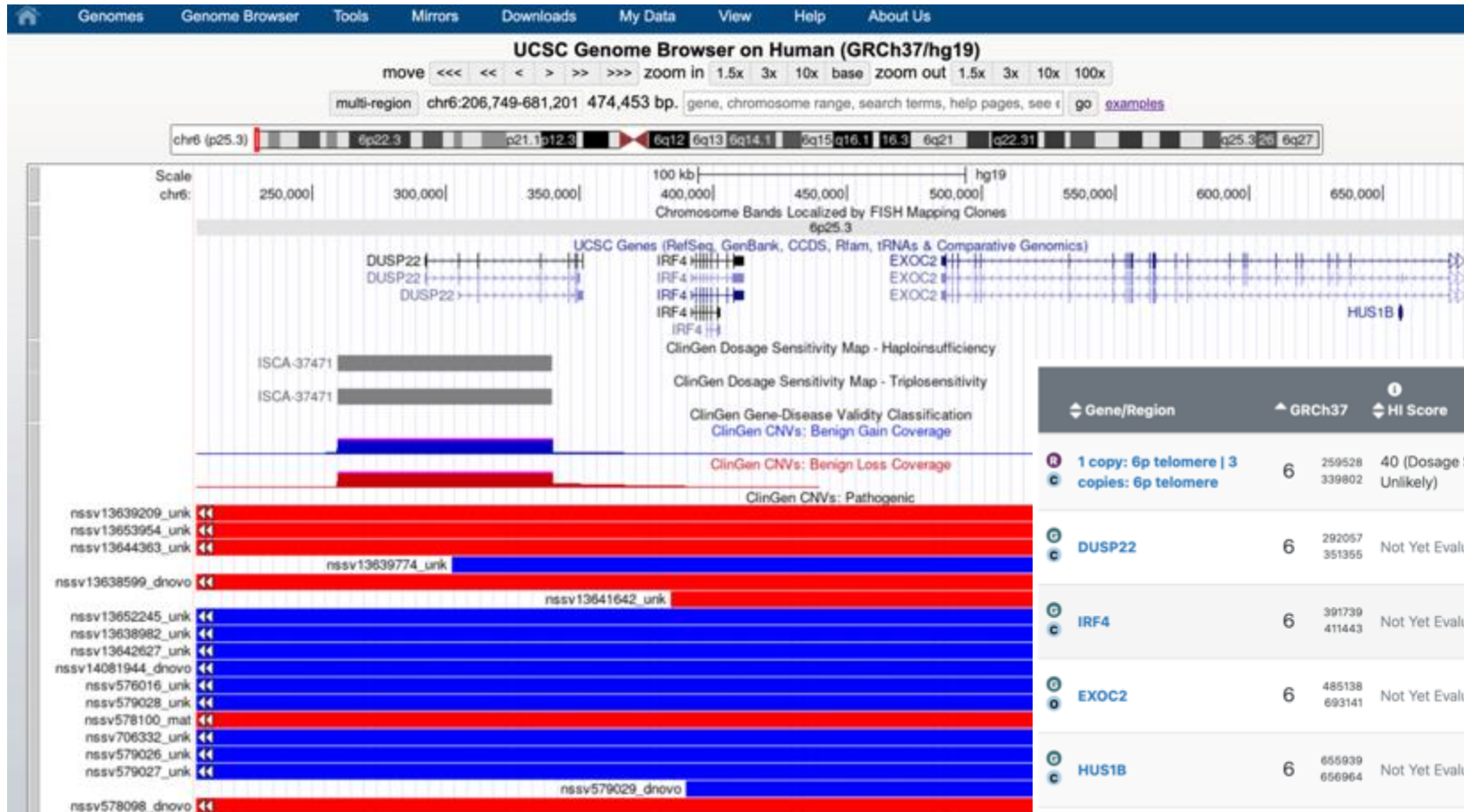
神奈川県立こども医療センター 遺伝科

黒田 友紀子

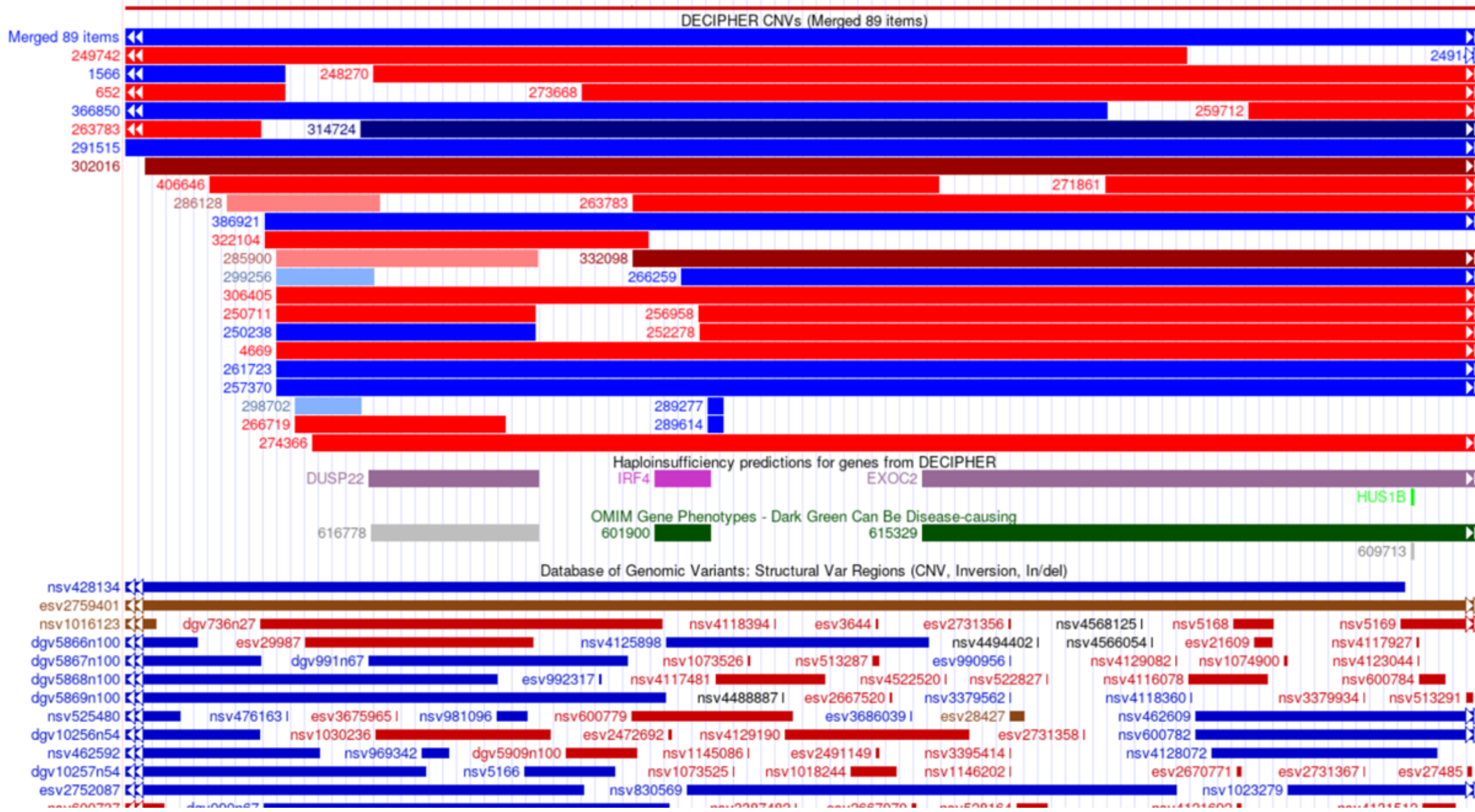
# 症例4

- 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



dgv5901n100

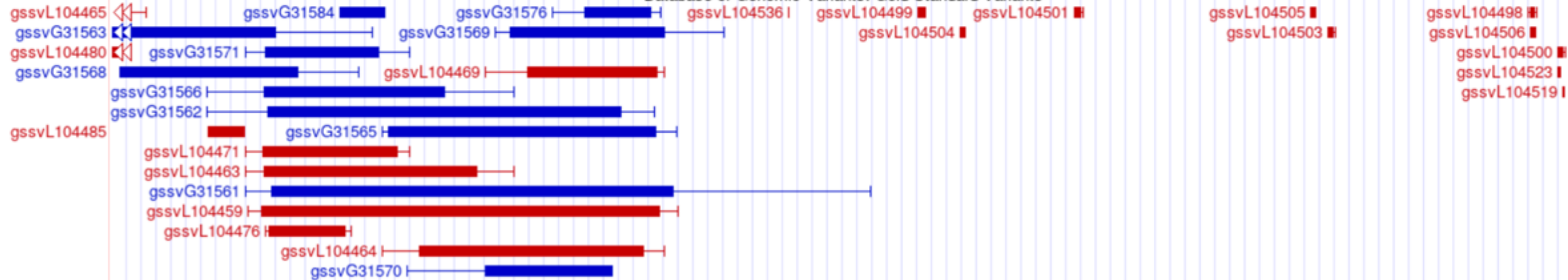
nsv1111687 |  
nsv1153784 |  
nsv3389871 |  
esv3567124 |  
nsv1121338 |  
nsv4114982 |

nsv3386875 |

Database of Genomic Variants: Structural Variation (CNV, Inversion, In/del)

DGV Supp Var

Database of Genomic Variants: Gold Standard Variants



RepeatMasker

Repeating Elements by RepeatMasker

Duplications of >1000 Bases of Non-RepeatMasked Sequence

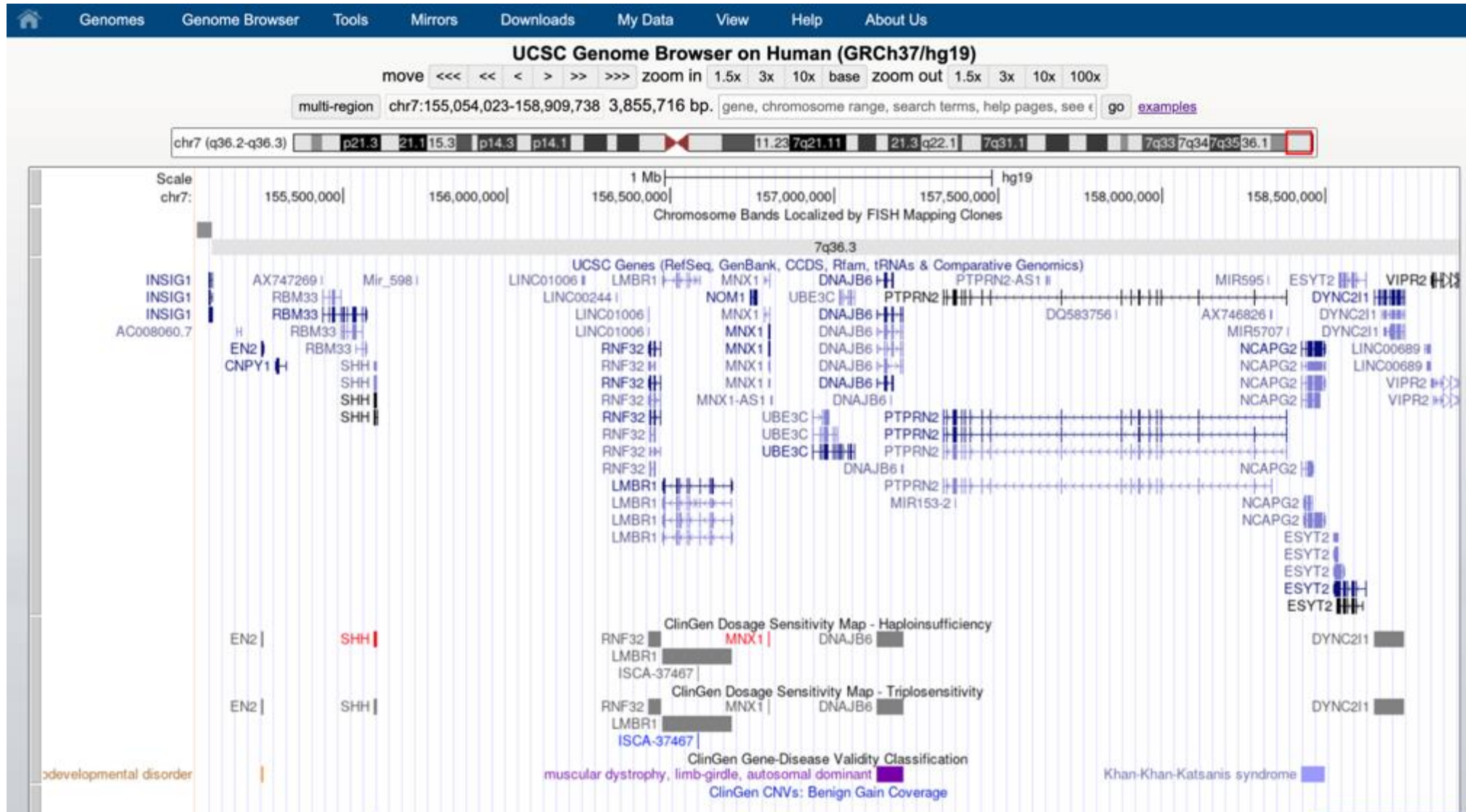
Segmental Dups

move start

Click on a feature for details. Shift+click/drag to zoom in. Click gray side bars for track options. Drag side bars or labels up or down to

Click to alter the display density of Segmental Dups and

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



Clonal 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  
nssv579057\_unk  
nssv578224\_unk

nssv578235\_unk

nssv706407\_unk  
nssv578163\_unk  
nssv579059\_unk  
nssv578218\_unk  
nssv578219\_unk  
nssv578222\_unk  
nssv578223\_unk

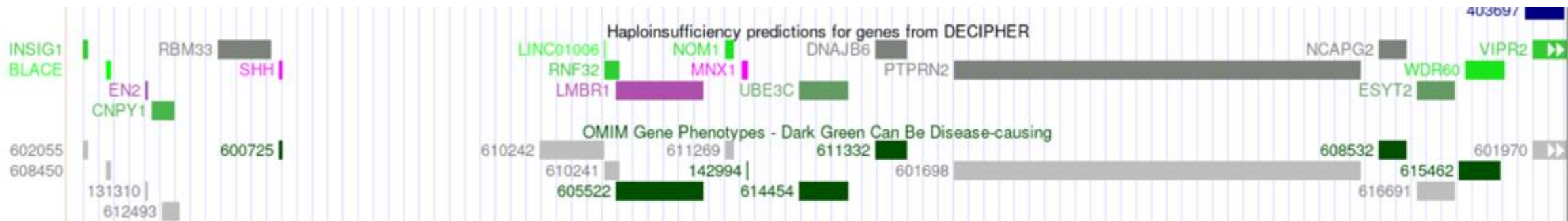
nssv578163\_unk

nssv578229\_dnov  
nssv578230\_dnov  
nssv578231\_dnov  
nssv578232\_unk  
nssv582774\_unk  
nssv582786\_dnov

nssv578236\_unk

nssv707022\_unk  
nssv707148\_unk

nssv706784\_unk



Database of Genomic Variants: Structural Var Regions (CNV, Inversion, In/del)

nsv609121	nsv476652	esv6152	nsv366031	nsv465248	nsv4481492	nsv6022	esv32984	nsv365287	esv21952	esv995097	esv7545	esv22916	nsv18
nsv4162785	nsv4524267	nsv6018	nsv4168882	esv27576	esv3615678	nsv4157667	esv25887	nsv8244	esv4996	esv28383	esv4054	esv5280	nsv6029
dgv11744n54	nsv3416098	nsv518485	esv24315	nsv609170	nsv4482427	nsv4162987	nsv4160565	nsv8243	esv3409	nsv6025	esv5759	esv25694	nsv951684
esv2003709	nsv4549128	nsv477572	esv2735620	nsv4163469	esv3672192	nsv4154404	nsv4158844	nsv8245	esv8571	nsv6026	esv27556	esv3311	esv28310
dgv6766n100	nsv4155626	nsv609164	nsv4557673	nsv442081	nsv4496609	nsv4161318	nsv366429	esv33838	esv26619	esv5555	esv4791	esv4476	nsv4525305
nsv1132109	nsv1144205	nsv509228	nsv4169196	nsv609177	nsv4166945	nsv4484464	nsv366618	nsv6023	nsv609260	esv25551	nsv4161512	nsv1027593	
esv3542739	esv2735587	nsv366255	nsv821124	esv2735638	esv3615676	esv3689019	nsv4168708	nsv365377	nsv951672	esv3956	nsv510974	nsv6028	
esv2670811	nsv4158421	esv2735592	nsv4545205	nsv365771	esv2668849	esv3615682	esv992340	nsv366775	nsv3411275	esv4672	esv991388	esv29148	
dgv2006e212	esv28471	esv4022	nsv4495790	nsv609174	esv2676333	esv3615683	nsv951670	nsv8246	esv2735825	nsv6027	nsv609363		
esv2671394	esv3401073	esv990012	nsv609169	esv991608	esv3542770	nsv4158805	nsv509229	nsv8249	esv3684	esv6030	esv23992	nsv465262	
nsv4162328	nsv3417071	nsv510971	nsv525273	nsv6019	nsv1146313	nsv4154496	esv7246	nsv8248	nsv4169270	esv29708	nsv970983	nsv8250	
esv2761389	nsv3408248	esv3677650	nsv831197	nsv4167969	nsv1137319	nsv4157536	nsv509230	esv28985	esv5043	nsv1032050	esv27462		
nsv1017558	nsv955996	nsv4156455	nsv4344904	esv2735644	nsv4165466	nsv966918	esv5094	esv22520	nsv509232	esv25118	nsv951681	nsv3401138	
nsv472867	nsv3406529	esv3677645	nsv3412937	nsv4488028	esv3615679	esv2665104	nsv1126823	esv23204	esv2370566	nsv951677	nsv438013	esv28360	
esv3615627	nsv3406898	esv3310459	dgv4008e59	nsv957049	esv2660160	nsv4160722	esv2735690	esv26337	nsv4162746	nsv442294	nsv4164687		
nsv519304	nsv3416571	esv3367702	nsv4495658	esv3679342	nsv4163604	nsv4153190	nsv8242	esv996958	nsv821497	esv8193	nsv824425	nsv3403349	
nsv609124	nsv3400392	esv2735604	nsv4162832	esv3689007	nsv1145171	esv2735670	nsv442286	nsv609248	esv23469	esv25476	nsv609419		
nsv609125	nsv609162	esv3542750	nsv1151409	nsv3404397	nsv4162715	esv2735671	nsv609212	esv3677705	nsv609293	nsv951679	esv27406	nsv366254	
esv3688996	esv3679316	nsv3418182	esv2735632	nsv4480685	esv3677660	nsv4154009	nsv119555	nsv366306	esv3615718	esv33849	nsv609364	esv9128	
nsv480657	nsv3404971	nsv1118032	dgv3719n106	nsv511377	esv3689010	nsv1117782	nsv428191		esv26378	esv275466	nsv951683		
nsv981620	nsv3409582	nsv3414041	nsv1076632	nsv3409858	nsv4162473	esv3677672	esv3677686	esv2735765	esv28090	esv3560	nsv4163998	esv2621732	
nsv476010	esv2735589	nsv957017	nsv4152770	nsv8241	nsv4524541	nsv3402187	dgv4014e59	esv2119297	nsv365303	nsv958461	nsv4487105	esv1955405	
nsv4555798	esv3677641	esv1192043	esv1505757	nsv4478574	esv2735652	nsv4343949			esv2736012	nsv609421			
esv3451864	nsv3408420	esv3677652	esv3417326	esv3542765	nsv1015433	esv3615684	esv3677687	esv987843	nsv512931	dgv138e19	nsv4495760	nsv3410881	
nsv4160244	esv2677211	esv996845	esv1410174	esv7116	esv3672193	dgv1347e214	esv2663923	nsv4525414	nsv1075494	nsv609358	esv3689063		
nsv609126	esv2656919	nsv1075490	nsv3417370	nsv512008	nsv3409181	nsv3404096	esv3542784	esv3615710	nsv4169560	nsv511365	nsv957631	nsv3411975	
nsv1144204	nsv3412124	nsv1137317	nsv3409712	esv2661583	nsv4158345	esv2735673	nsv4169937	nsv1110813	nsv4171547	nsv951680	nsv518319		
esv3679267	nsv1032585	nsv465246	nsv4551967	nsv6020	nsv824415	nsv951668	nsv4152481	esv2735782	esv2430415	nsv4170345	nsv4524633		
nsv1142507	nsv4489170	nsv512007	esv1512392	esv21970	nsv4343283	nsv1129792	esv3615703	esv2735770	nsv4166803	esv23376	nsv366535	nsv3414924	
nsv1115036	nsv4161257	esv8949	esv2735633	esv3689008	esv3677664	nsv1147621	nsv4163420	esv1951528	nsv1034867	nsv509237	nsv609423		
nsv1143197	nsv4153748	esv3664	esv3615656	esv6524	esv2735653	nsv1143199	nsv3415780	esv3677708	esv2735876	nsv509236	nsv527634		
nsv3416000	nsv2689000	esv508403	nsv4168222	esv2615667	esv2735654	esv2735674	esv2009850	esv2735771	esv4165459	nsv2402700	nsv2690065		



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		gssvL120400		gssvL120476	gssvL120613	gssvL120765	gssvL120940		
gssvL120246		gssvL120337		gssvL120489	gssvL120616	gssvL120770	gssvG36177		
gssvL120247		gssvL120339		gssvL120492	gssvL120618	gssvL120758	gssvL120935		
gssvL120259		gssvL120340		gssvL120509	gssvL120622	gssvL120757	gssvL120939		
gssvL120265		gssvL120346	gssvL120414	gssvL120512	gssvL120626	gssvG36247	gssvL120942		
gssvL120263		gssvL120357	gssvL120415	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%

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）	<i>DUSP22, IRF4, EXOC2</i> 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 <i>SHH, MNX1</i> 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)	

