Fichiers BEDs

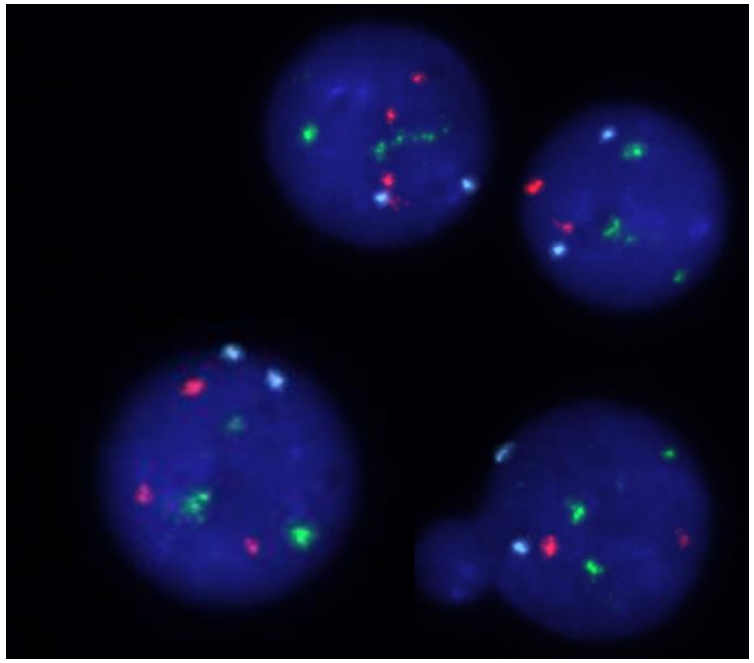
- Sondes FISH (Metasystem)
- DGV gold standard

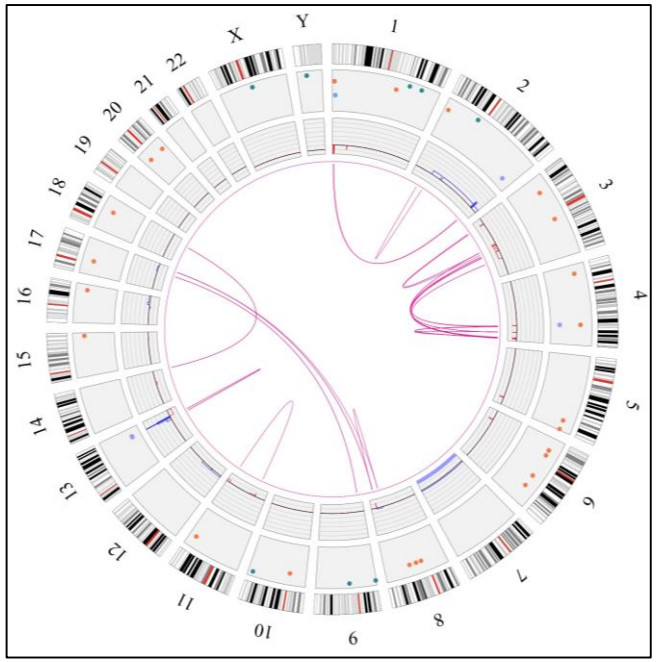
24/06/2025

Sondes FISH

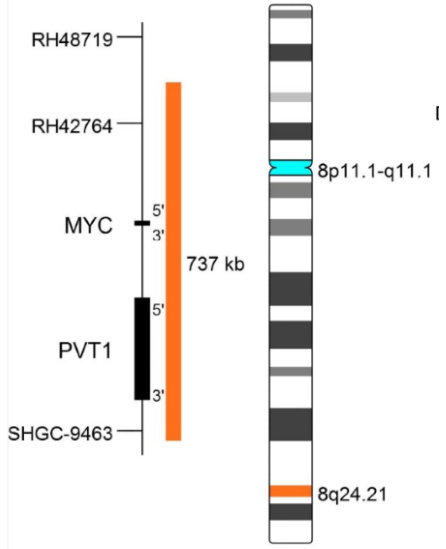
- Réalisation d'un fichier recensant les positions des sondes FISH utiles en cytogénétique hémato au CHU de LILLE (en Hg38)
- Travail initié par Margot DUCOURANT (externe en pharmacie CHU Lille)

→ **Faciliter l'interprétation conjointe des dossiers caryo/FISH/bionano**

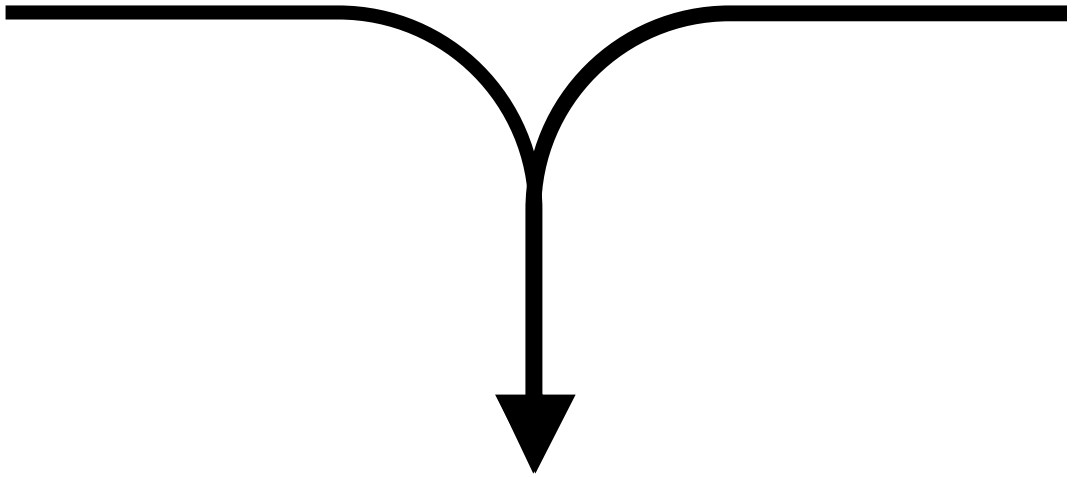


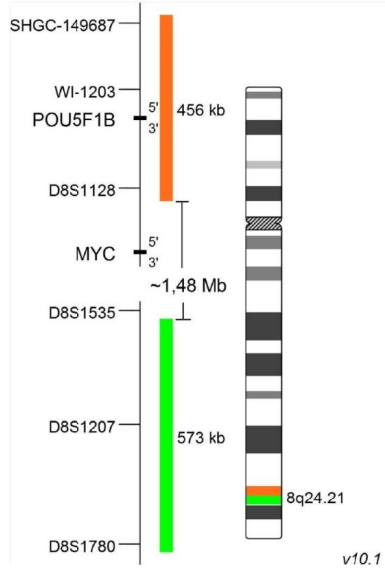


Sondes FISH

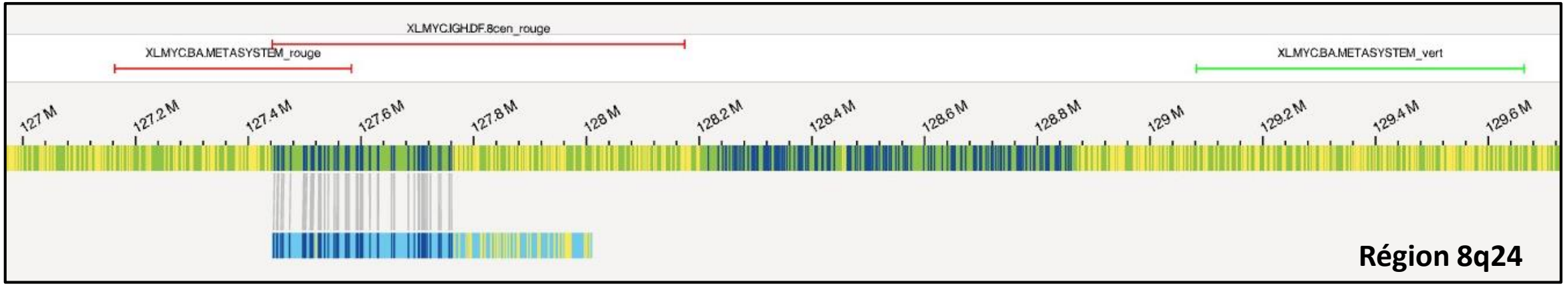


XL t(8;14) MYC-IGH-8cen
METASYSTEM



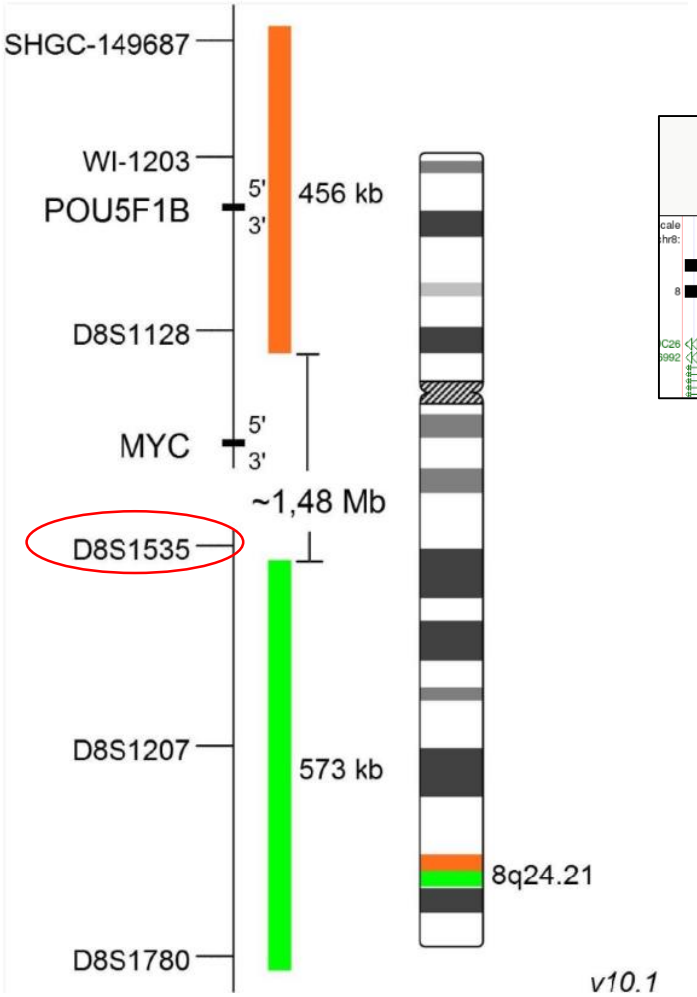


XL MYC BA
METASYSTEM



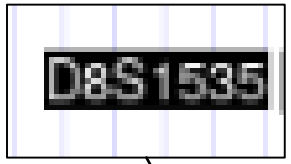
Région 8q24

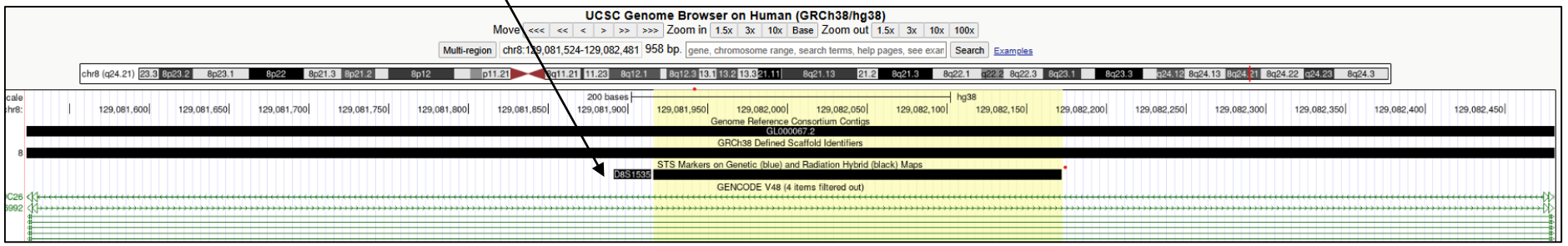
Sondes FISH

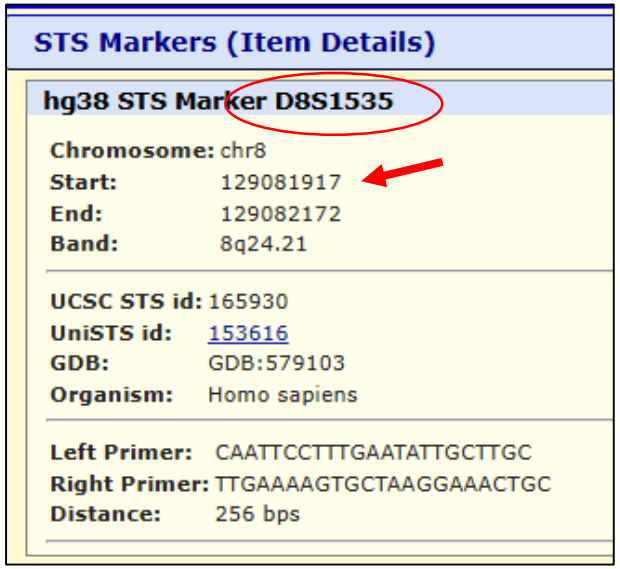


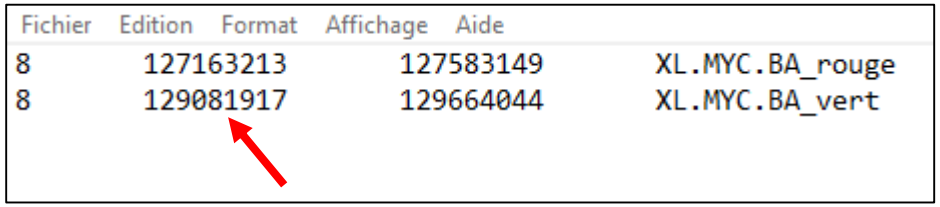
XL MYC BA

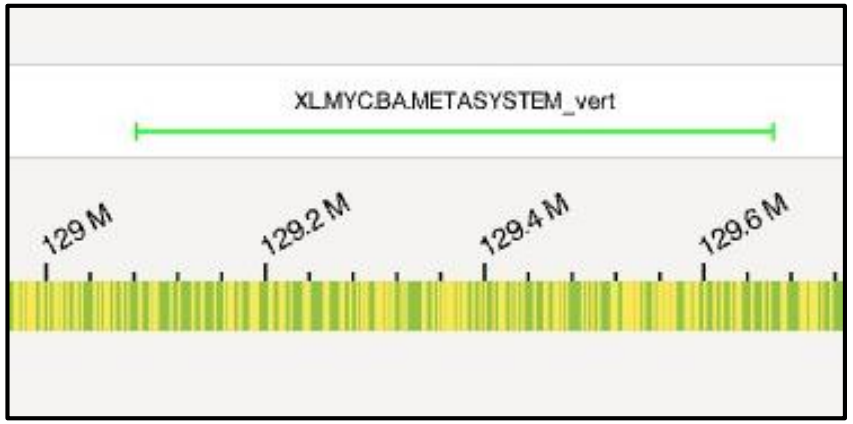
METASYSTEM











Sondes FISH

Aperçu du ficher

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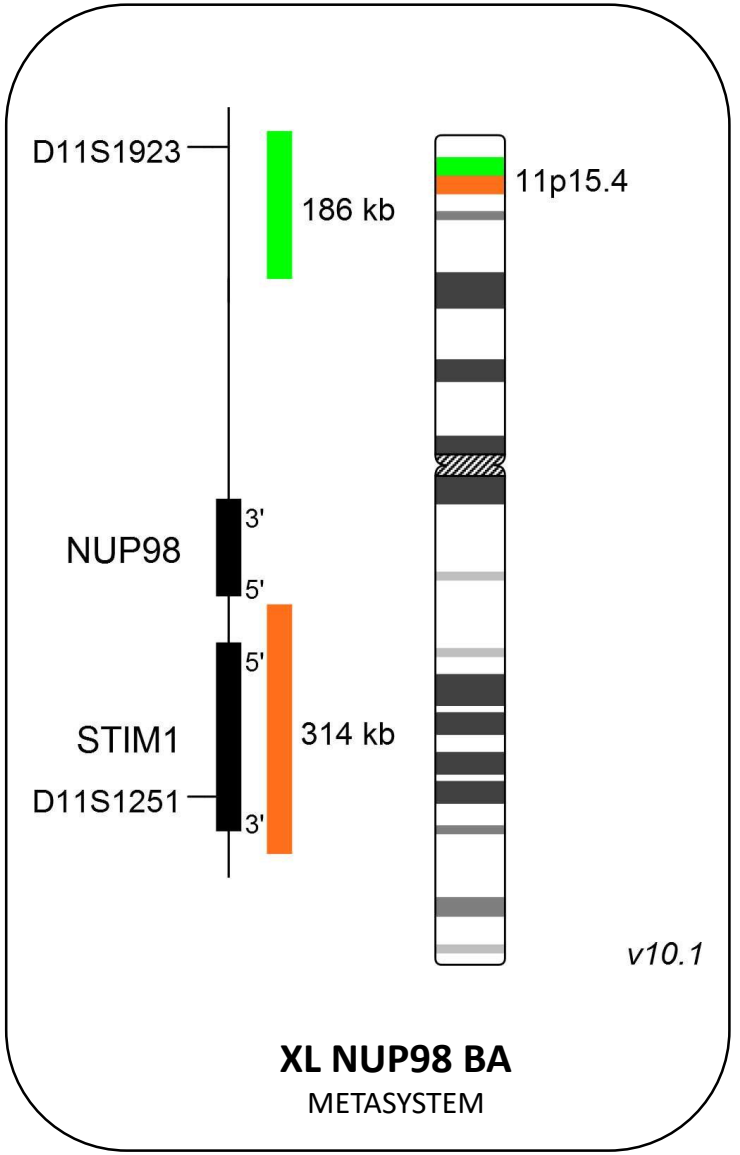
2	151121335	151448990	XL MYCN amp METASYSTEM10				255,0,0
2	88186288	88557015	XL IGK BA METASYSTEM11				0,255,0
2	88570742	88750742	XL IGK BA METASYSTEM12				0,255,0
2	89885706	90228706	XL IGK BA METASYSTEM13				255,0,0
3	187078222	187251615	XL BCL6 BA METASYSTEM14				0,255,0
3	187335997	187670394	XL BCL6 BA METASYSTEM15				0,255,0
3	188055742	188469617	XL BCL6 BA METASYSTEM16				255,0,0
3	168566222	168696170	XL MECOM METASYSTEM17				255,0,0
3	168737079	168901079	XL MECOM METASYSTEM18				255,0,0
3	169342974	169501715	XL MECOM METASYSTEM19				0,255,0
3	169528787	169857521	XL MECOM METASYSTEM20				0,255,0
3	169938476	170072069	XL MECOM METASYSTEM21				0,255,0



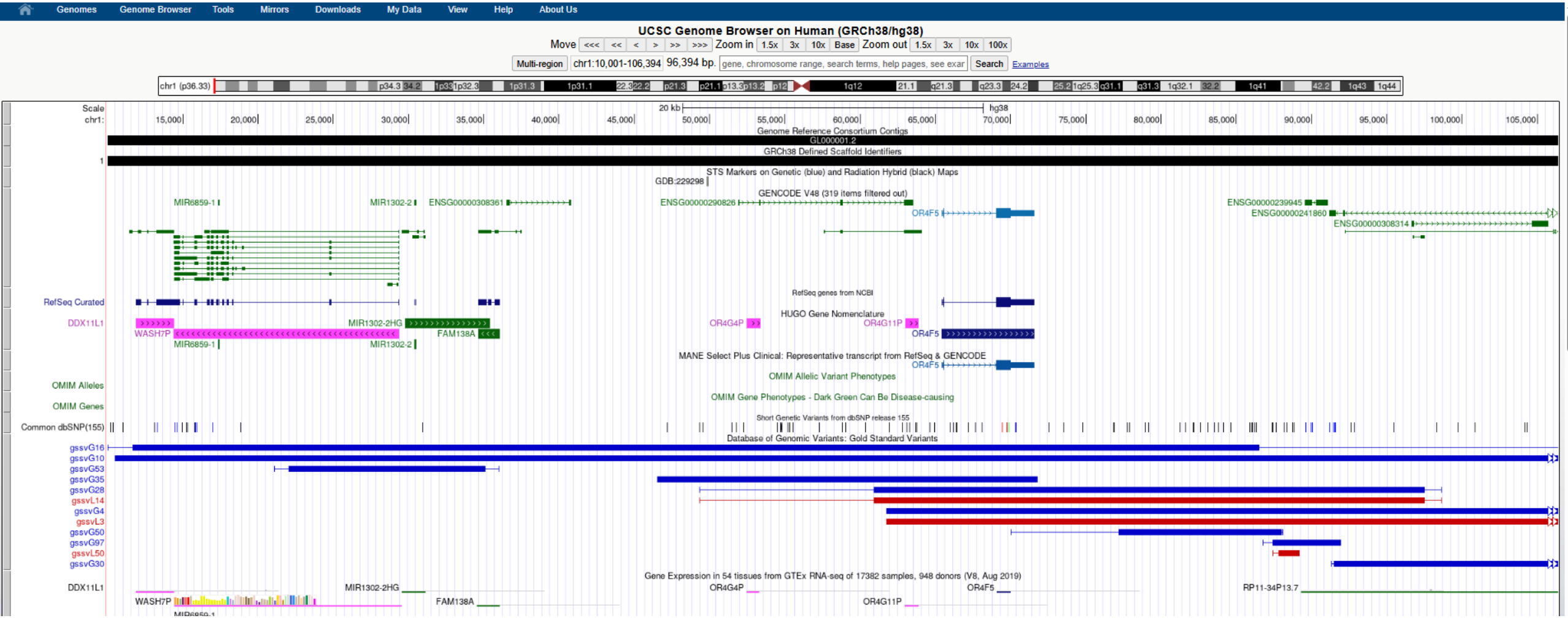
- BED pratique
- Précision des *loci* dépendante de la précision du schéma de sonde, parfois il persiste une inexactitude (ex. NUP98 BA)
- Principales sondes FISH Metasystem, le fichier peut être amélioré, précisé et agrémenté avec les sondes des autres fournisseurs
- Le chiffre après le fournisseur de sonde ne veut rien dire

→ Toutes erreurs ou incohérences observées à l'usage est utile à remonter

DGV gold standard

- Difficulté d'interpréter certains variants dans la *curated variant lists*
- L'anomalie parait convaincante (nombre de molécules, alignement, …) pour autant est-ce un polymorphisme non présent dans les patients « contrôle » de bionano ou est-ce un potentiel variant de la population de cellules cancéreuses ? La VAF peut ne pas être un critère de décision sur les échantillons très infiltrés
- Les indications de type « nombre de DGV calls » peuvent aider mais souvent le recours à la visualisation UCSC est nécessaire, pour observer la position des variants DGV et comparer à notre anomalie
- Le BED « DGV » de bionano est trop lourd pour l'ouvrir dans Access (à voir selon les centre)

DGV gold standard



DGV gold standard

Database of Genomic Variants

A curated catalogue of human genomic structural variation

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Download Data

The contents of the Database of Genomic Variants can be downloaded as tab delimited text files. The first line of each file is the column description. Each variant is provided with an accession which is a stable identifier and will remain constant. Studies where CNV coordinates are based on a newer assembly are not mapped back to older builds. The NCBI Build 36 (hg18) download file will therefore contain less data than the GRCh37 (hg19).

We have provided three categories of files for users to download. The first set of files, contained in the "DGV variants" section, represents the data that is displayed in our primary DGV structural variants track. The second set of files, in the "Supporting Variants" section, are the sample level and supporting variants that are displayed in our Supporting Variants track. The last section is the "Filtered Variants" files. These are variants that have been removed from the database following our curation process.

Note that for the DGV Variants and Supporting Variants sections, a new file has been made available under the "Other Mappings" column. These files contain those variants that do not map to the reference assembly but instead to an alternate sequence/contig assembly.

DGV Variants

Release Date	Build 36 (hg18)	GRCh 37 (hg19)	GRCh 38 (hg38)	Other Mappings
2020-02-25	link	link	link	
2016-05-15	link	link	link	
2015-07-23	link	link	link	
2014-10-16	link	link	link	
2013-07-23	link	link		link
2013-05-31	link	link		link

Supporting Variants

Release Date	Build 36 (hg18)	GRCh 37 (hg19)	GRCh 38 (hg38)	Other Mappings
2020-02-25	link	link	link	
2016-05-15	link	link	link	
2015-07-23	link	link	link	
2014-10-16	link	link	link	
2013-07-23	link	link		link
2013-05-31	link	link		link

Filtered Variants

Release Date	Build 36 (hg18)	GRCh 37 (hg19)	GRCh 38 (hg38)
2016-02-25	link	link	link
2016-05-15	link	link	link
2015-07-23	link	link	link
2014-10-16	link	link	link

DGV Gold Standard Variants

Release Date	GRCh 37 (hg19)	GRCh 38 (hg38)
2016-05-15	link	link
2015-07-23	link	

Old versions (note that each file contains both DGV Variants and Supporting Variants)

Release Date	Build 36 (hg18)	GRCh 37 (hg19)	Other Mappings
2012-11-14	link	link	link

DGV gold standard


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chr1	CNV	copy_number_variation_region	49298	49298	.	.	.
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ID=gssvL14;Name=gssvL14;variant_type=CNV;variant_sub_type=Loss;outer_start=49298;inner_start=60905;inner_end=97505;outer_end=98602;inner_rank=14;num_variants=16;variants=essv6990744,essv6991855,essv7004857,essv7004858,essv7004859,essv7004860,essv7004861,essv7004864,essv7004869,essv7004871,essv7004872,essv7004873,essv7027856,essv7028967,nssv707617,essv6990742;num_studies=2;Studies=Vogler2010,Cooper2011;num_platforms=2;Platforms=Affymetrix6.0,Custom_Illumina_1.2M;number_of_algorithms=2;algorithms=PennCNV+Birdseye,Custom_HMM;num_samples=16;samples=RW_0620,RW_0502,RW_0608,SW_0892,SW_1265,SW_1273,RW_0611,SW_0257,RW_0166,RW_0064,RW_0148,sample_nssv707617,RW_0077,SW_0148,RW_0137,RW_0071;Frequency=0.613262%;PopulationSummary=African 10:European 6:Asian 0:Admixed 0:Mexican 0:MiddleEast 0:Unknown 0:NativeAmerican 0:NorthAmerican 0:Oceania 0:SouthAmerican 0:Turkish 0;Number_of_unique_samples_tested=2609
chr1	CNV	copy_number_variation_region	60905	97505	.	.	.
ID=gssvG28;Name=gssvG28;variant_type=CNV;variant_sub_type=Gain;outer_start=49298;inner_start=60905;inner_end=97505;outer_end=98602;inner_rank=14;num_variants=16;variants=essv6992966,essv6994077,essv7001854,essv7004862,essv7004863,essv7004865,essv7004866,essv7004868,essv7004870,essv7012965,essv7024076,essv7030078,essv7031189,essv7032300,nssv707616,essv6990743;num_studies=2;Studies=Vogler2010,Cooper2011;num_platforms=2;Platforms=Affymetrix6.0,Custom_Illumina_1.2M;number_of_algorithms=2;algorithms=PennCNV+Birdseye,Custom_HMM;num_samples=16;samples=SW_1092,SW_1361,RW_0564,RW_0229,SW_1425,SW_1000,RW_0233,SW_1085,SW_0831,RW_0155,RW_0161,sample_nssv707616,SW_0060,SW_0044,SW_0086,RW_0184;Frequency=0.613262%;PopulationSummary=African 6:European 10:Asian 0:Admixed 0:Mexican 0:MiddleEast 0:Unknown 0:NativeAmerican 0:NorthAmerican 0:Oceania 0:SouthAmerican 0:Turkish 0;Number_of_unique_samples_tested=2609
chr1	CNV	copy_number_variation_region	60905	97505	.	.	.
ID=gssvL14;Name=gssvL14;variant_type=CNV;variant_sub_type=Loss;outer_start=49298;inner_start=60905;inner_end=97505;outer_end=98602;inner_rank=14;num_variants=16;variants=essv6990744,essv6991855,essv7004857,essv7004858,essv7004859,essv7004860,essv7004861,essv7004864,essv7004869,essv7004871,essv7004872,essv7004873,essv7027856,essv7028967,nssv707617,essv6990742;num_studies=2;Studies=Vogler2010,Cooper2011;num_platforms=2;Platforms=Affymetrix6.0,Custom_Illumina_1.2M;number_of_algorithms=2;algorithms=PennCNV+Birdseye,Custom_HMM;num_samples=16;samples=RW_0620,RW_0502,RW_0608,SW_0892,SW_1265,SW_1273,RW_0611,SW_0257,RW_0166,RW_0064,RW_0148,sample_nssv707617,RW_0077,SW_0148,RW_0137,RW_0071;Frequency=0.613262%;PopulationSummary=African 10:European 6:Asian 0:Admixed 0:Mexican 0:MiddleEast 0:Unknown 0:NativeAmerican 0:NorthAmerican 0:Oceania 0:SouthAmerican 0:Turkish 0;Number_of_unique_samples_tested=2609
chr1	CNV	copy_number_variation_region	61723	61723	.	.	.
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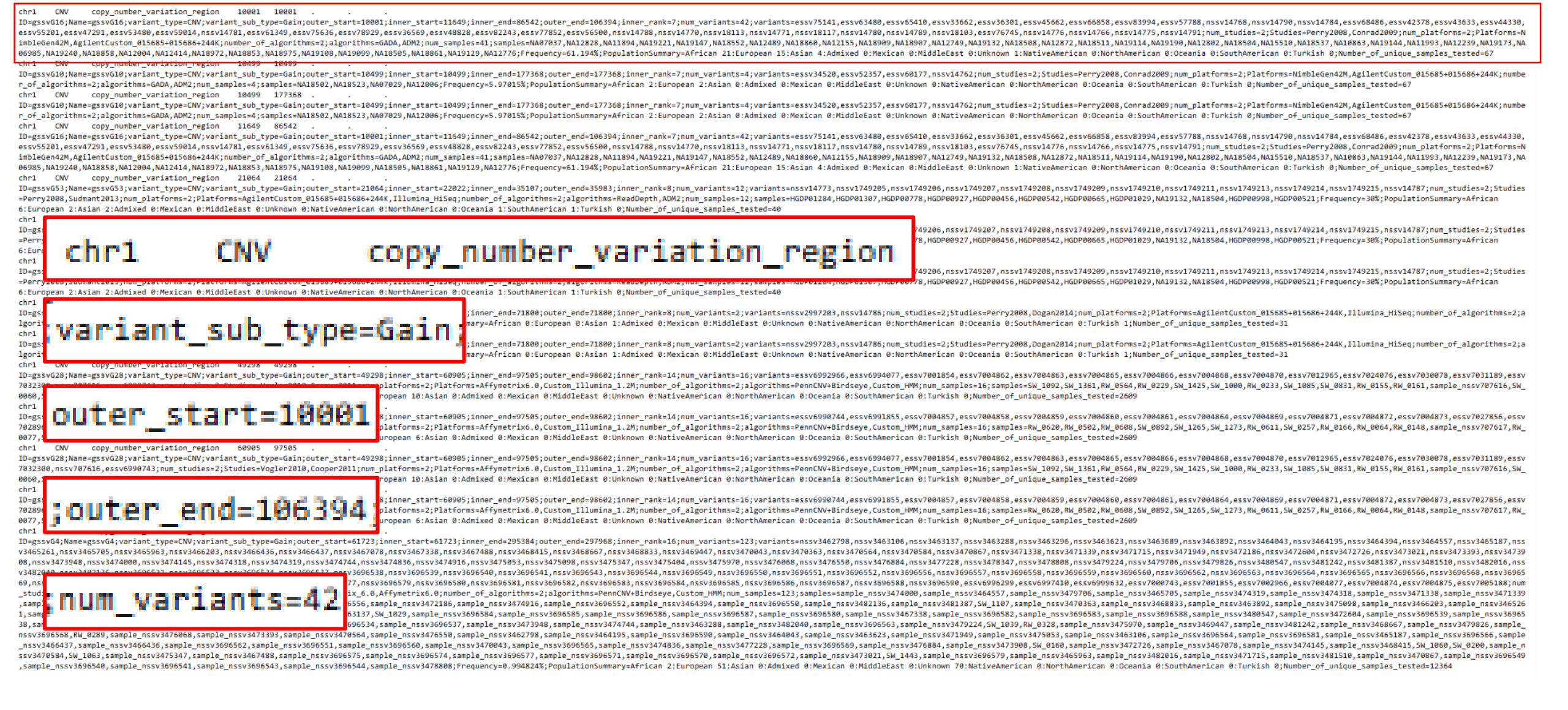
Liste très longue à charger, page à dérouler jusqu'en bas…

DGV gold standard



DGV gold standard

DGV Gold Standard (Item Details)

hg38 Human: Database of Genomic Variants: Gold Standard Variants (gssvG16)

DGV Browser and Report: gssvG16
Item: gssvG16
Position: chr1:10001-106394
Band: 1p36.33
Genomic Size: 96394
View DNA for this feature (hg38/Human)

Name of CNV from DGV	gssvG16
Variant Type	CNV
Gain or Loss	Gain
Rank of the CNV used to assign the blocks	7
Number of supporting variants	42
Supporting variants	essv75141, essv63480, essv65410, essv33662, essv36301, essv45662, essv66858, essv83994, essv57788, nssv14768, nssv14790, nssv14784, essv68486, essv42378, essv43633, essv44330, essv55201, essv47291, essv53480, essv59014, nssv14781, essv61349, essv75636, essv78929, essv36569, essv48828, essv82243, essv77852, essv56500, nssv14788, nssv14770, nssv18113, nssv14771, nssv18117, nssv14780, nssv14789, nssv18103, essv76745, nssv14776, nssv14766, nssv14775, nssv14791
Number of studies	2
Study names in 'Name Year' format	Perry2008, Conrad2009
Number of platforms	2
Platform names	NimbleGen42M, AgilentCustom_015685+015686+244K
Number of CNV detection algorithms	2
CNV detection algorithms used	GADA, ADM2
Number of samples	41
Sample names	NA07037, NA12828, NA11894, NA19221, NA19147, NA18552, NA12489, NA18860, NA12155, NA18909, NA18907, NA12749, NA19132, NA18508, NA12872, NA18511, NA19114, NA19190, NA12802, NA18504, NA15510, NA18537, NA10863, NA19144, NA11993, NA12239, NA19173, NA06985, NA19240, NA18858, NA12004, NA12414, NA18972, NA18853, NA18975, NA19108, NA19099, NA18505, NA18861, NA19129, NA12776
Overall frequency of variants across all studies	61.194%
Populations tested across all studies	African 21, European 15, Asian 4, Admixed 0, Mexican 0, MiddleEast 0, Unknown 1, NativeAmerican 0, NorthAmerican 0, Oceania 0, SouthAmerican 0, Turkish 0
Total number of samples tested	67

Close

DGV gold standard

DGV Gold Standard (Item Details)

hg38 Human: Database of Genomic Variants: Gold Standard Variants (gssvG16)

DGV Browser and Report: gssvG16
Item: gssvG16
Position: chr1:10001-106394
Band: 1p36.33
Genomic Size: 96394
View DNA for this feature (hg38/Human)

Name of CNV from DGV	gssvG16
Variant Type	CNV
Gain or Loss	Gain
Rank of the CNV used to assign the blocks	7
Number of supporting variants	42
Supporting variants	essv75141, essv63480, essv65410, essv53480, essv59014, nssv14781, nssv18103, essv76745, nssv14776, ... essv55201, essv47291, ... ssv14780, nssv14789,
Number of studies	2
Study names in 'Name Year' format	Perry2008, Conrad2009
Number of platforms	2
Platform names	NimbleGen42M, AgilentCustom_015
Number of CNV detection algorithms	2
CNV detection algorithms used	GADA, ADM2
Number of samples	41
Sample names	NA07037, NA12828, NA11894, NA1 ... NA18537, NA10863, NA19144, NA1 ... 02, NA18504, NA15510, ... 29, NA12776
Overall frequency of variants across all studies	61.194%
Populations tested across all studies	African 21, European 15, Asian 4, Admixed 0, Mexican 0, MiddleEast 0, Unknown 1, NativeAmerican 0, NorthAmerican 0, Oceania 0, SouthAmerican 0, Turkish 0
Total number of samples tested	67

DGV_gold_standard_all_g__PR_bis - Bloc-notes

Fichier Edition Format Affichage Aide

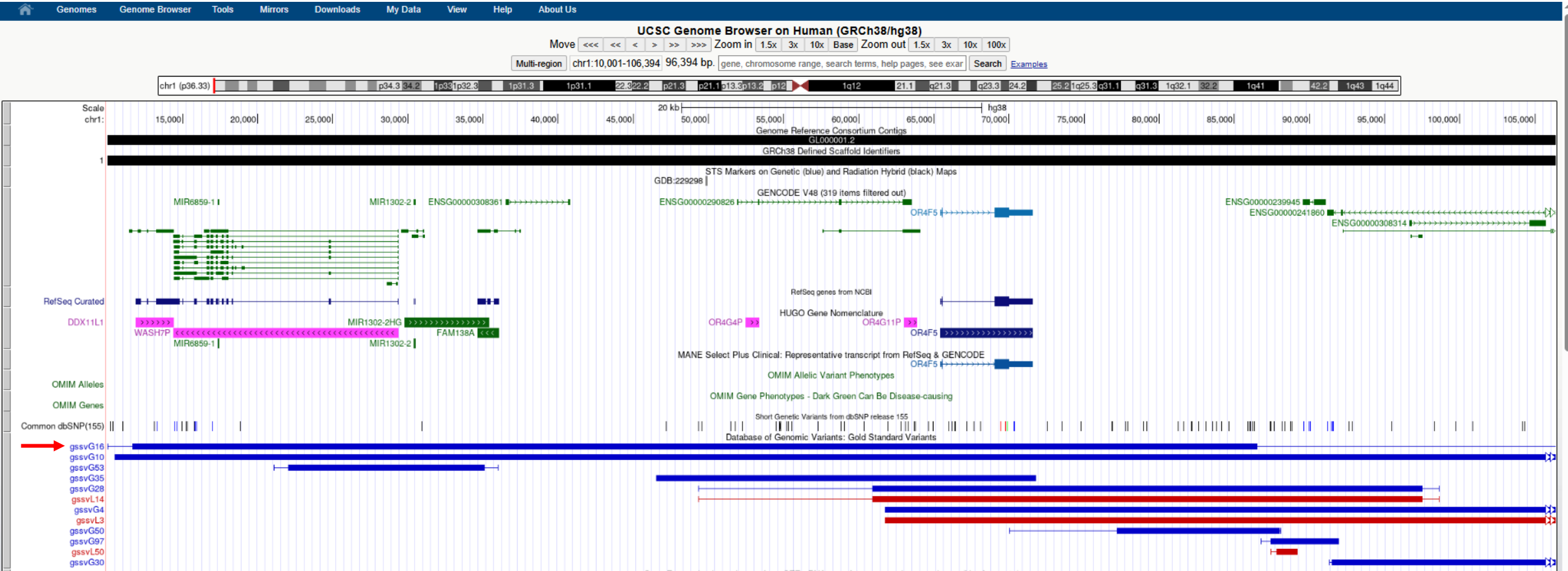
1	10001	106394	gssvG16_42	0,0,255
1	10499	177368	gssvG10_4	0,0,255
1	21064	35983	gssvG53_12	0,0,255
1	46501	71800	gssvG35_2	0,0,255
1	49298	98602	gssvG28_16	0,0,255
1	49298	98602	gssvL14_16	255,0,0
1	61723	297968	gssvG4_123	0,0,255
1	61723	297968	gssvL3_37	255,0,0
1	70007	88084	gssvG50_11	0,0,255
1	86778	91967	gssvG97_13	0,0,255
1	87433	89163	gssvL50_2	255,0,0
1	91317	125400	gssvG30_11	0,0,255
1	121042	176984	gssvG24_11	0,0,255
1	136181	137811	gssvL51_5	255,0,0

Close

DGV gold standard

Fichier Edition Format Affichage Aide

1	10001	106394	gssvG16_42	0,0,255
1	10499	177368	gssvG10_4	0,0,255
1	21064	35983	gssvG53_12	0,0,255
1	46501	71800	gssvG35_2	0,0,255

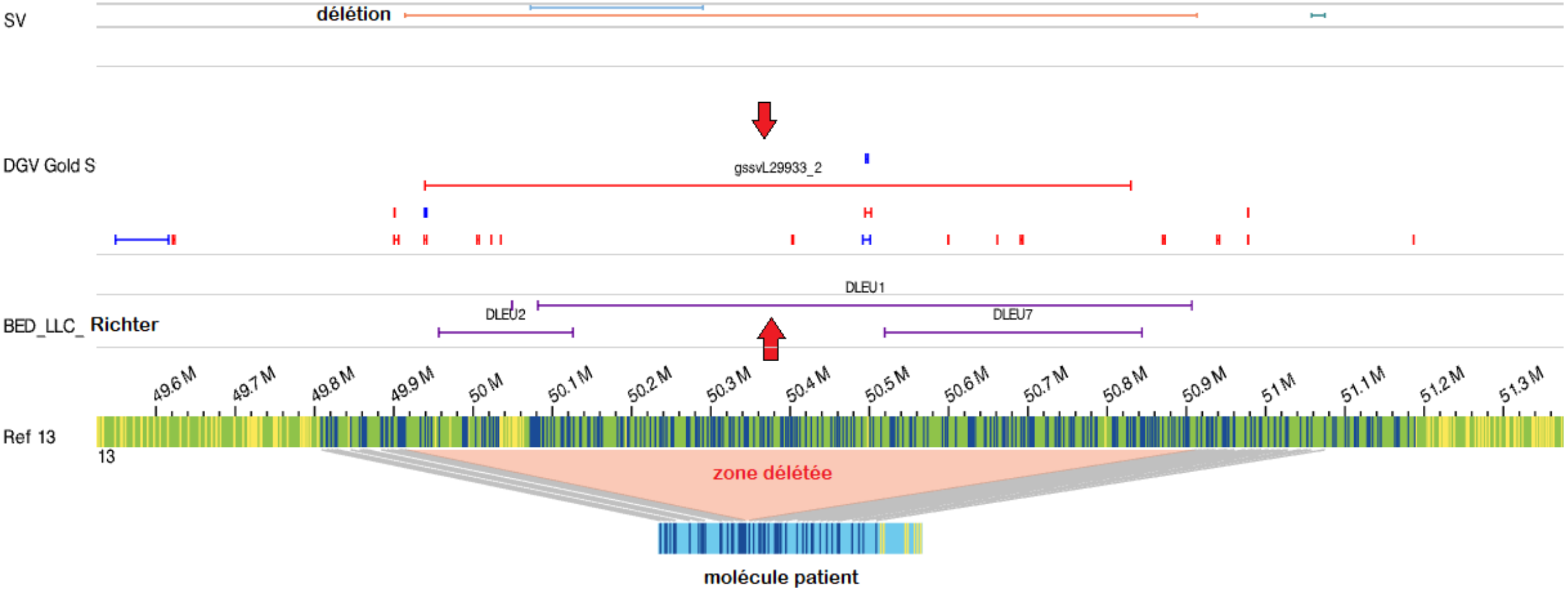


DGV gold standard

Visuel Bionano

DGV gold standard

Attention !



DGV gold standard

A savoir sur ce bed

Fichier Edition Format Affichage Aide

1	10001	106394	gssvG16_42	0,0,255
1	10499	177368	gssvG10_4	0,0,255
1	21064	35983	gssvG53_12	0,0,255
1	46501	71800	gssvG35_2	0,0,255
1	49298	98602	gssvG28_16	0,0,255
1	49298	98602	gssvL14_16	255,0,0
1	61723	297968	gssvG4_123	0,0,255
1	61723	297968	gssvL3_37	255,0,0
1	70007	88084	gssvG50_11	0,0,255
1	86778	91967	gssvG97_13	0,0,255
1	87433	89163	gssvL50_2	255,0,0
1	91317	125400	gssvG30_11	0,0,255
1	121042	176984	gssvG24_11	0,0,255
1	136181	137811	gssvL51_5	255,0,0

- Couleur pour gain (bleu ; 0,0,255) vs délétion (rouge ; 255,0,0)
- Borne externe répertoriée (vis-à-vis du visuel UCSC)
- Nom du variant DGV cf. diapo suivante

DGV gold standard

- Nom = nom du DGV gold standard variant puis « _ » et number of supporting variants, parfois à l'état homozygote chez un sujet donc ne veut pas dire nombre de patients avec ce variant, et pas d'idée sur le nombre de sujets étudiés total

Fichier Edition Format Affichage Aide

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1       10001   106394  gssvG16_42                      0,0,255
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DGV Gold Standard (Item Details)

hg38 Human: Database of Genomic Variants: Gold Standard Variants (gssvG16)

DGV Browser and Report: gssvG16
Item: gssvG16
Position: chr1:10001-106394
Band: 1p36.33
Genomic Size: 96394
View DNA for this feature (hg38/Human)

Name of CNV from DGV	gssvG16
Variant Type	CNV
Gain or Loss	Gain
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Number of studies	2
Study names in 'Name Year' format	Perry2008, Conrad2009
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Overall frequency of variants across all studies	61.194%
Populations tested across all studies	African 21, European 15, Asian 4, Admixed 0, Mexican 0, MiddleEast 0, Unknown 1, NativeAmerican 0, NorthAmerican 0, Oceania 0, SouthAmerican 0, Turkish 0
Total number of samples tested	67

Close

Fichiers à disposition pour le FrOGG

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DGV_gold_standard_VF.bed	20/06/2025 16:37	Fichier BED	1 760 Ko