Bazy danych wykorzystywane w analizie wyników badań cytogenetycznych

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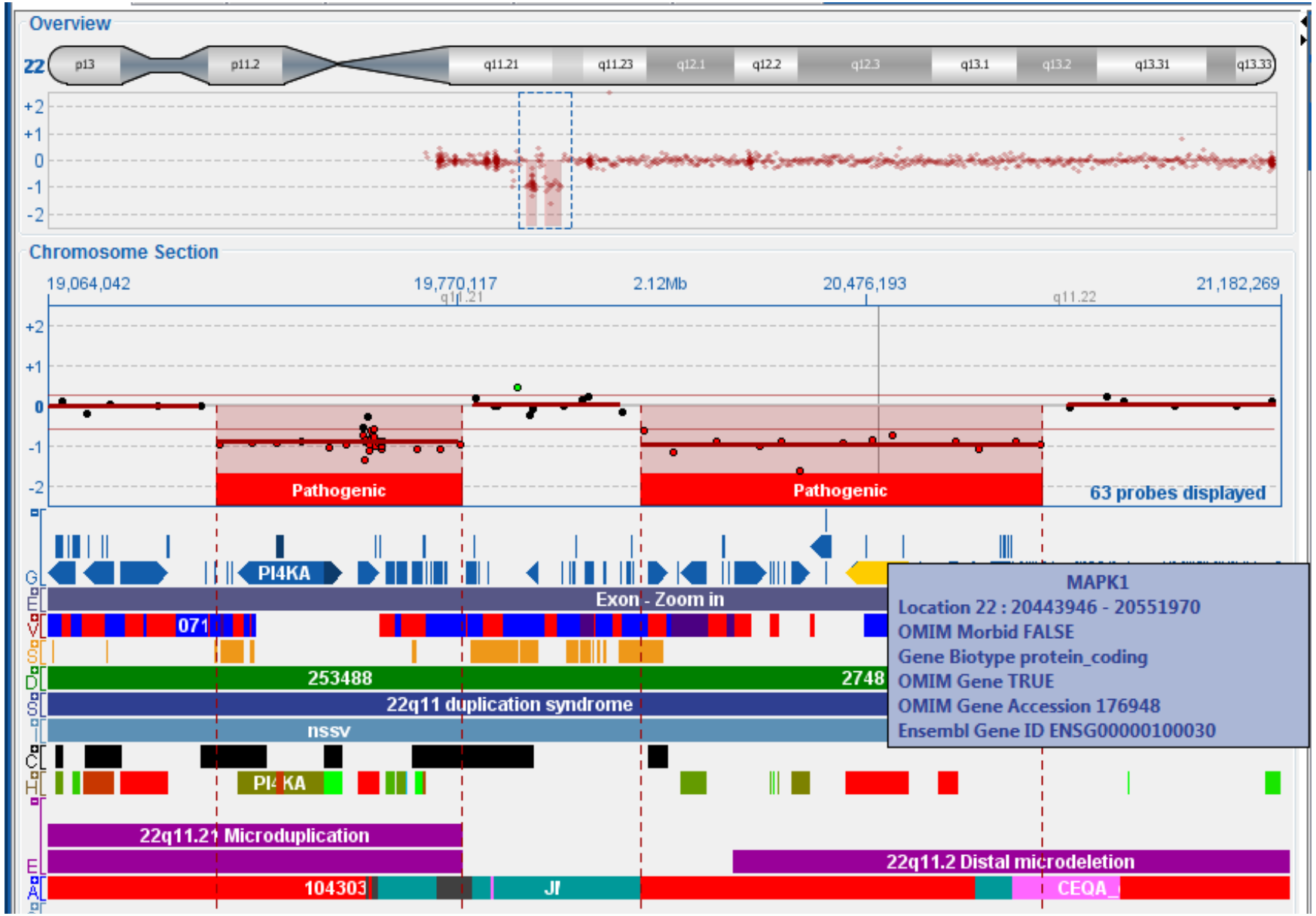
Rzeczpospolita Polska
Instytut Matki i Dziecka
Unia Europejska Europejski Fundusz Społeczny
edu-metgen.imid.med.pl

Table 2. List of Online Resources Used in CMA Results Interpretation.		
Name	**Abbreviation**	**Website**
Database of Genomic Variants	DGV	http://projects.tcag.ca/variation
Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources	DECIPHER	https://decipher.sanger.ac.uk/application
Database of Genotype and phenotype	dbGaP	http://www.ncbi.nlm.nih.gov/gap
Database of Structural Variation	dbVar	http://www.ncbi.nlm.nih.gov/dbvar
European Cytogeneticists Association Register of Unbalanced Chromosome Aberrations	ECARUCA	http://www.ECARUCA.net
Genomic Oligoarray and SNP array evaluation tool		http://www.ccs.miami.edu/cgi-bin/ROH/ROH_analysis_tool.cgi
International Standards for Cytogenomic Arrays Consortium	ISCA	https://www.iscaconsortium.org
NCBI Nucleotide		http://www.ncbi.nlm.nih.gov/nuccore
NCBI Online Mendelian Inheritance in Man	OMIM	http://www.ncbi.nlm.nih.gov/omim
NCBI PubMed	PubMed	http://www.ncbi.nlm.nih.gov/PubMed
USCS Genome Browser	UCSC	http://genome.ucsc.edu/cgi-bin/hgGateway
Prenatal Array		http://prenatalarray.org

CytoSure Oxford Gene Technology


Overview
22
p13
p11.2
q11.21
q11.23
q12.1
q12.2
q12.3
q13.1
q13.2
q13.31
q13.33
+2
+1
0
-1
-2
Chromosome Section
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19,770,117
2.12Mb
20,476,193
21,182,269
q11.21
q11.22
Pathogenic
Pathogenic
63 probes displayed
PI4KA
Exon - Zoom in
071
253488
2748
22q11 duplication syndrome
nssv
PI4KA
22q11.21 Microduplication
22q11.2 Distal microdeletion
104303
JI
CEQA_
MAPK1
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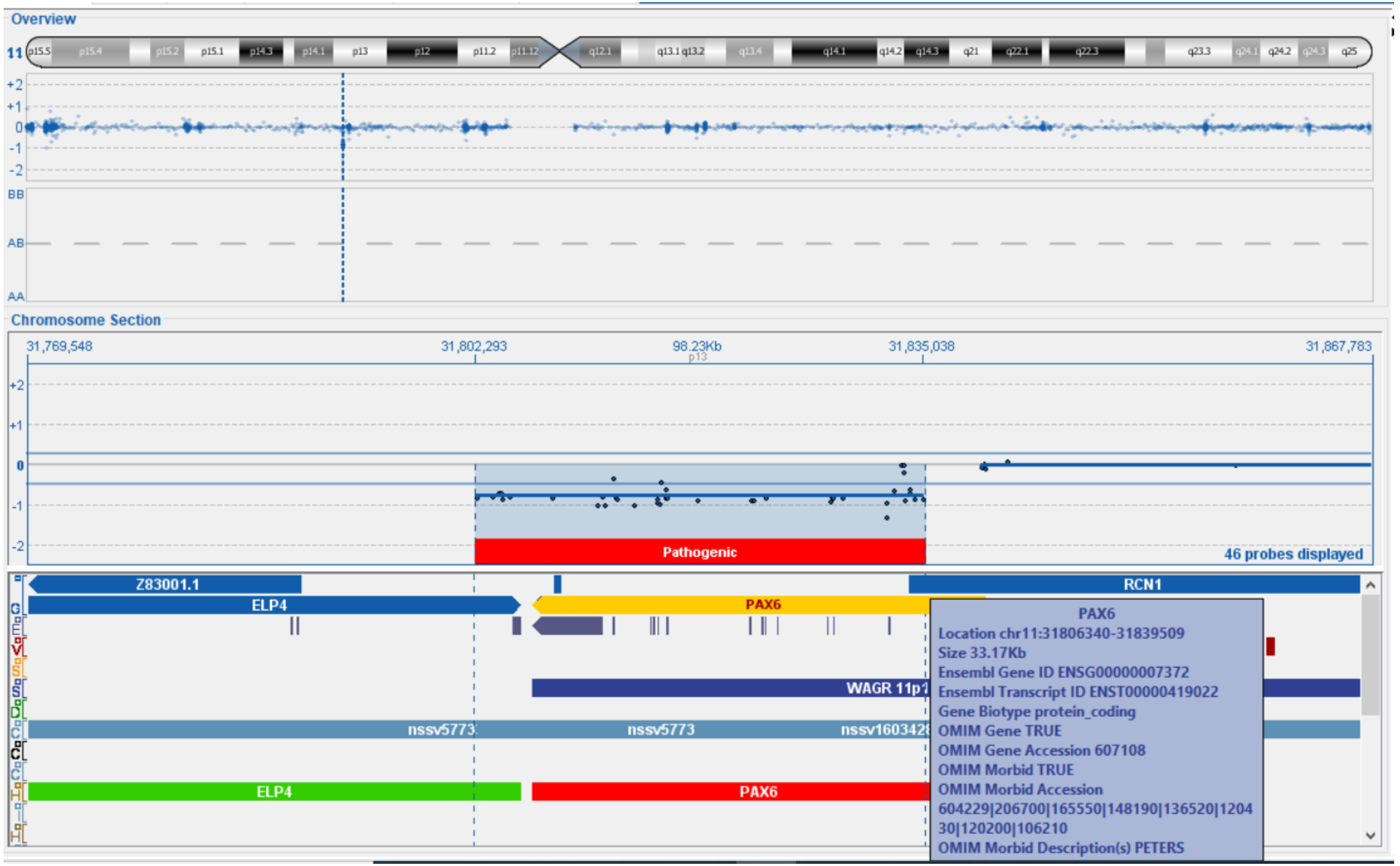
OMIM Morbid FALSE
Gene Biotype protein_coding
OMIM Gene TRUE
OMIM Gene Accession 176948
Ensembl Gene ID ENSG00000100030

GENY


Overview
11
p15.5 p15.4 p15.2 p15.1 p14.3 p14.1 p13 p12 p11.2 p11.12 q12.1 q13.1 q13.2 q13.4 q14.1 q14.2 q14.3 q21 q22.1 q22.3 q23.3 q24.1 q24.2 q24.3 q25
+2
+1
0
-1
-2
BB
AB
AA
Chromosome Section
31,769,548
31,802,293
98.23Kb
p13
31,835,038
31,867,783
Pathogenic
46 probes displayed
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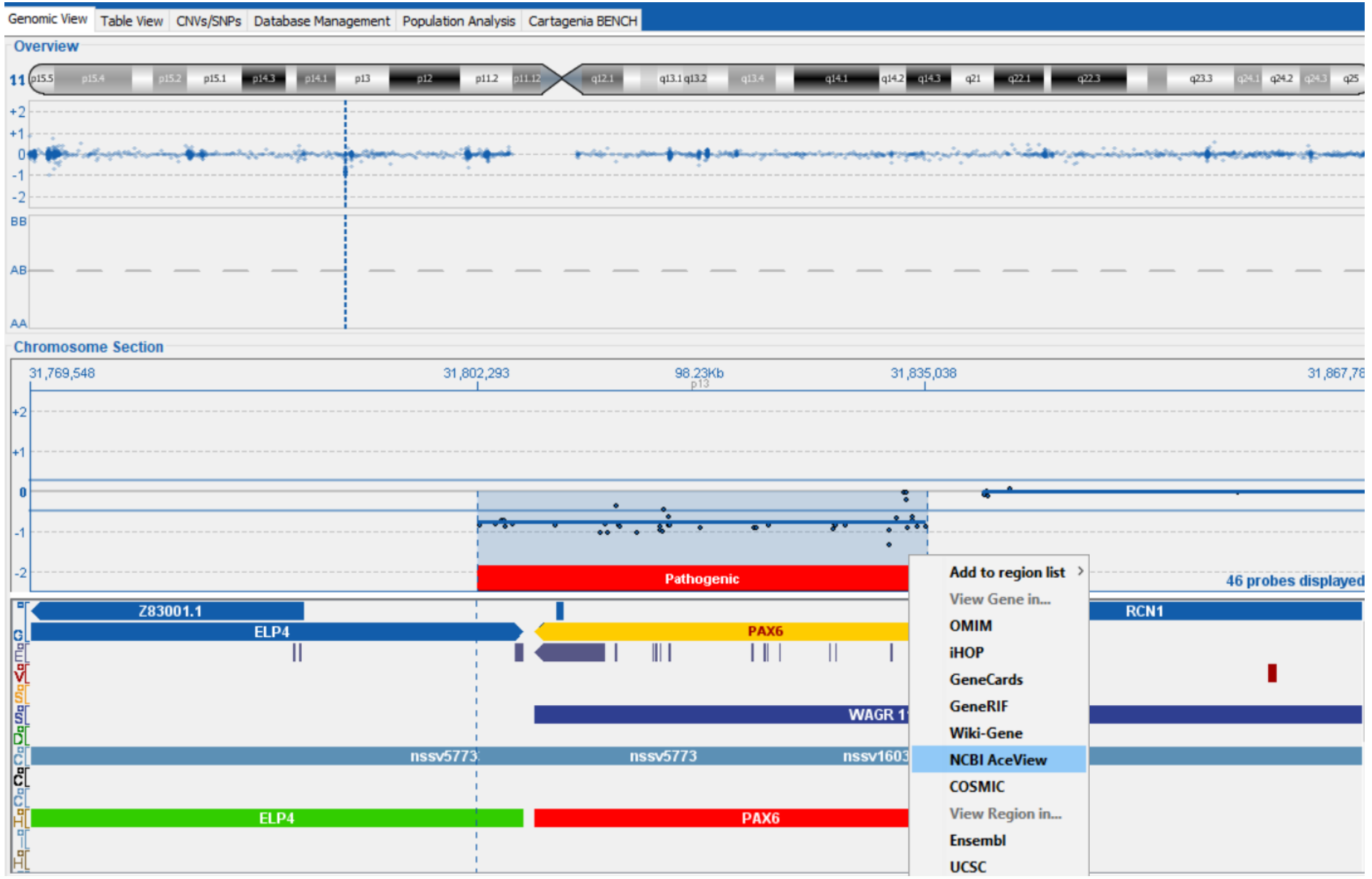
RCN1
ELP4
PAX6
WAGR 11p1
nssv5773
nssv5773
ELP4
PAX6
PAX6
Location chr11:31806340-31839509
Size 33.17Kb
Ensembl Gene ID ENSG00000007372
Ensembl Transcript ID ENST00000419022
Gene Biotype protein_coding
OMIM Gene TRUE
OMIM Gene Accession 607108
OMIM Morbid TRUE
OMIM Morbid Accession
604229|206700|165550|148190|136520|120430|120200|106210
OMIM Morbid Description(s) PETERS



Informacje o genach z regionu aberracji

https://www.ncbi.nlm.nih.gov/IEB/Research/Acembly/av.cgi?db=human&term=PAX6&submit=Go

NCBI

AceView

[Help on Query](#)

Select a species

in Human 2010

[Search by name](#)

e.g: PTEN, FGF, ada, diabetes, mitotic spindle, AF369653*

Go

[Search by sequence](#)

Blast AceView

[News](#)

[FAQ](#)

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[Publications](#)

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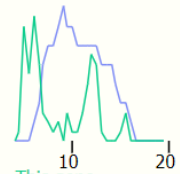
? | Gene Summary | Gene on genome | mRNA:.a, .b, .c, .d, .e, .f, .g, .h, .i, .j, .k, .l, .m, .n, .o, .p-u, .q, .r-u, .s, .t, .u-u, .va, .vb, .vc, .vd-u, .ve, .vf, .vg, .vh, .vi-u | Alternative mRNAs features, proteins, introns, exons, sequences | Expression Tissue | Function, regulation, related genes DCI

PAX6 — Gene expression in 15 primates, 16 tissues, from the NHPRTR project in sFPKM

	HUM	CHP	PTM	JMI	RMI	RMC	CMM	CMC	BAB	SMY	MST	SQM	OWL	MLM	RTL
Cerebellum		5.40	43.2	11.6	32.8		43.2	43.2	40.3	99.3		28.5		53.2	
Brain	18.8	2.05	4.39	7.64	4.71		4.10	5.40	3.33	5.79	3.82	4.10		6.65	
Pituitary		2.05	4.10	2.90	3.33		6.21	5.79	4.71	6.21	6.21				
Ovary	2.70														
Testis	2.90				0.72										
Kidney	2.70		0.06		0.07			0.05							
Liver	0.96									0.04				0.05	
Heart	1.45		0.10	0.05	0.05						0.68	0.06			
SkeletalMuscle	0.83									0.05	0.04				
Lung	0.78				0.07				0.06	0.04	0.06			0.08	
Colon	1.91	0.07	0.27	0.11			0.18	0.32	0.36	0.34	0.10	0.32		0.10	
BoneMarrow		0.06	0.09					0.07	0.09	0.04	0.07				
Spleen			0.07	0.06											
LymphNode	0.96	0.06	0.13	0.06							0.10				
Thymus		0.18	0.13	0.08	0.10		0.06	0.07	0.06	0.18					
WholeBlood	1.55														

Expression quantiles: None, Weak, 1, 2, 3, 4, 5, 6, 7, 8



RNA-seq gene expression profile across 16 selected tissues from the Non-Human Primates Reference Transcriptome Resource (link to NHPRTR project).
- Primates: Apes (HUM: Human (Illumina BodyMap 2), CHP: Chimpanzee), Old World monkeys (PTM: Pig-Tailed Macaque, JMI Japanese Macaque, RMI Rhesus Macaque Indian, RMC Rhesus Macaque Chinese, CMM Cynomolgus Macaque Mauritian, CMC Cynomolgus Macaque Chinese, BAB Olive Baboon, SMY Sooty Mangabey); New World monkeys (MST common Marmoset, SQM Squirrel Monkey, OWL Owl Monkey); and Lemurs (MLM Mouse Lemur, RTL Ring-Tailed Lemur).
- The level for significantly expressed genes is color coded in 8 equal sized bins (light to dark green). Light gray is for weak not-accurately measured expression (2 to 8 reads above intergenic background); dark gray for no expression or no sequence conservation (0 read in gene).
The plot to the right shows the distribution of measured expression values in all tissues for all genes (blue) and for this gene (green), in Magic index = $\log_2$(1000 sFPKM).

You may also examine the strand-specific genome coverage plots on the experimental AceView/Magic hub at UCSC, by tissue or by species. Tracks may be slow to load; please reload if some tracks come up yellow-greenish, and thanks to UCSC for the great work!.

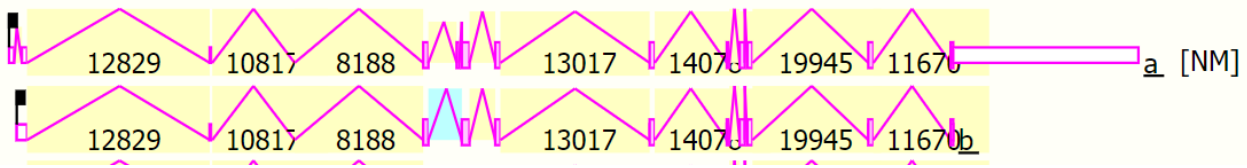
Read more...

▼ Complete gene on genome diagram:

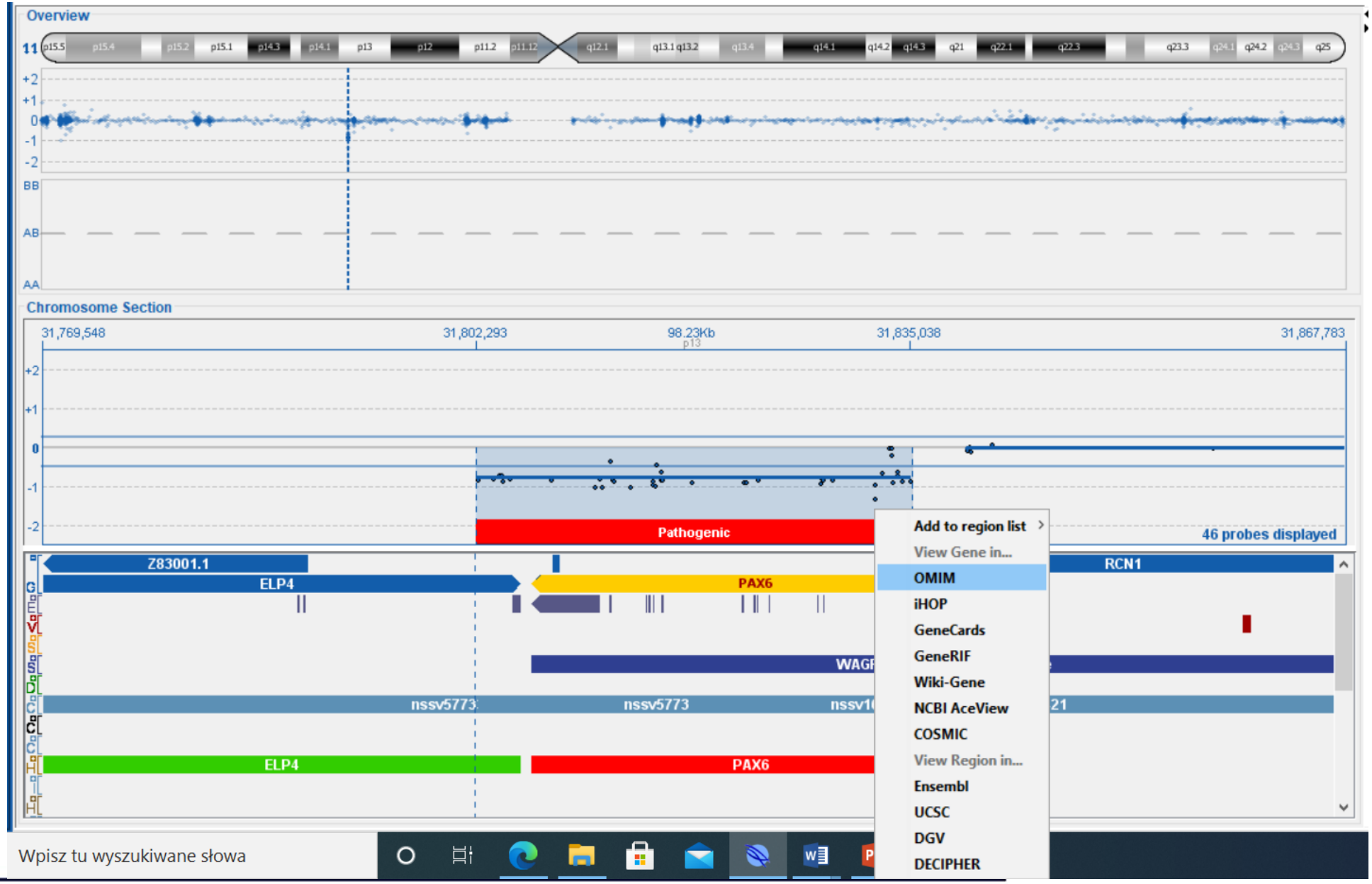
Please choose between the zoomable GIF version., and the HTML5/SVG version.
This diagram shows in true scale the gene on the genome, the mRNAs and the cDNA clones.

▼ Compact gene diagram

Gene PAX6 5' → 3' encoded on minus strand of chromosome 11 from 31,839,495 to 31,804,903



Roczny chłopiec. Wskazanie: aniridia obustronna (zaburzenie rozwojowe polegające na braku tęczówki oka)
arr[hg19] 11p13(31,802,293-31,835,038)x1 o wielkości ~91 kb


Overview
11
p15.5 p15.4 p15.2 p15.1 p14.3 p14.1 p13 p12 p11.2 p11.12 q12.1 q13.1 q13.2 q13.4 q14.1 q14.2 q14.3 q21 q22.1 q22.3 q23.3 q24.1 q24.2 q24.3 q25
+2
+1
0
-1
-2
BB
AB
AA
Chromosome Section
31,769,548
31,802,293
98.23Kb
p13
31,835,038
31,867,783
Pathogenic
46 probes displayed
Z83001.1
ELP4
PAX6
RCN1
WAGR
nssv5773
nssv5773
ELP4
PAX6
Add to region list
View Gene in...
OMIM
iHOP
GeneCards
GeneRIF
Wiki-Gene
NCBI AceView
COSMIC
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Wpisz tu wyszukiwane słowa

Search OMIM... Options Display: Highlights

*607108
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* 607108

PAIRED BOX GENE 6; PAX6

HGNC Approved Gene Symbol: PAX6

Cytogenetic location: 11p13 *Genomic coordinates (GRCh38): 11:31,789,025-31,817,960* (from NCBI)

Gene-Phenotype Relationships

Location	Phenotype Clinical Synopses	Phenotype MIM number	Inheritance	Phenotype mapping key
11p13	?Coloboma of optic nerve	120430	AD	3
	?Coloboma, ocular	120200	AD	3
	?Morning glory disc anomaly	120430	AD	3
	Aniridia	106210	AD	3
	Anterior segment dysgenesis 5, multiple subtypes	604229	AD	3
	Cataract with late-onset corneal dystrophy	106210	AD	3
	Foveal hypoplasia 1	136520	AD	3
	Keratitis	148190	AD	3
	Optic nerve hypoplasia	165550	AD	3

External Links
- Genome
- DNA
- Protein
- Gene Info
- Clinical Resources
- Variation
  - 1000 Genome
  - ClinVar
  - gnomAD
  - GWAS Catalog
  - GWAS Central
  - HGMD
  - HGVS
  - Locus Specific DBs
  - NHLBI EVS
  - PharmGKB
- Animal Models

ClinVar

ClinVar | PAX6[gene] | Search

Create alert | Advanced | Help

Home | About | Access | Help | Submit | Statistics | FTP

COVID-19 Information

Public health information (CDC) | Research information (NIH) | SARS-CoV-2 data (NCBI) | Prevention and treatment information (HHS) | Español

SIDEBAR

Clinical significance
Conflicting interpretations (30)
Benign (63)
Likely benign (51)
Uncertain significance (110)
Likely pathogenic (48)
Pathogenic (289)
Risk factor (0)

Molecular consequence
Frameshift (106)
Missense (101)
Nonsense (60)
Splice site (48)
ncRNA (357)
Near gene (0)
UTR (296)

Variation type
Deletion (146)
Duplication (55)
Indel (10)
Insertion (54)
Single nucleotide (352)

Variant length
Less than 51 bp (507)
Between 51 and 1000 bp (2)

Tabular | 100 per page | Sort by Location | Download:

Showing for results for variants in the **PAX6** gene. *Search instead for all ClinVar records that mention **PAX6***

Search results

Items: 1 to 100 of 570

<< First | < Prev | Page 1 of 6 | Next > | Last >>

	Variation *Location*	Gene(s)	Protein change	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession
1.	GRCh38/hg38 11p15.1-13(chr11:20079474-34463996)x1 *GRCh37:* Chr11:20101020-34485543 *GRCh38:* Chr11:20079474-34463996	ABTB2, ANO3, ANO3-AS1, ANO5, ARL14EP, ARL14EP-DT, BBOX1, BBOX1-AS1, BDNF, BDNF-AS, C11orf91, CAPRIN1, CAT, CCDC179, CCDC34, CCDC73, CD59, CSTF3, CSTF3-DT, DBX1, DCDC1, DEPDC7, DNAJC24, EIF3M, ELP4, FANCF, FBXO3 ...more		See cases	Pathogenic (Aug 30, 2010)	no assertion criteria provided	VCV000154432
2.	GRCh38/hg38 11p14.3-12(chr11:22550115-38199159)x1 *GRCh37:* Chr11:22571661-38220709 *GRCh38:* Chr11:22550115-38199159	BDNF, MPPED2, CAT, CD44, CD59, CSTF3, ELF5, FANCF, FSHB, GAS2, KCNA4, LMO2, CAPRIN1, **PAX6**, RAG1, RAG2, RCN1, SLC1A2, TRAF6, WT1, PDHX, BBOX1, HIPK3, EIF3M, FJX1, KIAA1549L, ABTB2, PAMR1, FBXO3, EHF, ELP4, COMMD9, APIP ...more		See cases	Pathogenic (Aug 12, 2011)	criteria provided, single submitter	VCV000058858
3.	GRCh38/hg38 11p14.2-13(chr11:26368962-35252976)x1 *GRCh37:* Chr11:26390509-35274523 *GRCh38:* Chr11:26368962-35252976	BDNF, MPPED2, CAT, CD44, CD59, CSTF3, ELF5, FSHB, KCNA4, LMO2, CAPRIN1, **PAX6**, RCN1, SLC1A2, WT1, PDHX, BBOX1, HIPK3, EIF3M, KIAA1549L, ABTB2, FBXO3, EHF, ELP4, APIP, WT1-AS, NAT10, LIN7C, TCP11L1, LGR4, ANO3, PRRG4, OSER1 ...more		See cases	Pathogenic (Nov 30, 2010)	no assertion criteria provided	VCV000145969

Frameshift (72)
Missense (0)
Nonsense (6)
Splice site (10)
ncRNA (83)
Near gene (0)
UTR (55)

Variation type clear
✓ **Deletion** (146)
Duplication (0)
Indel (10)
Insertion (10)
Single nucleotide (0)

Variant length
Less than 51 bp (99)
Between 51 and 1000 bp (2)
Between 1 and 50 kb (7)
Between 50 and 500 kb (4)
Between 500 kb and 1 Mb (0)
Between 1 and 5 Mb (0)
Greater than 5 Mb (3)

Review status
Practice guideline (0)
Expert panel (0)
Multiple submitters (4)
Single submitter (71)
At least one star (75)
Conflicting interpretations (0)

Allele origin
Germline (60)
De novo (27)
Somatic (0)

Method type
Research (6)
Literature only (1)
Clinical testing (60)

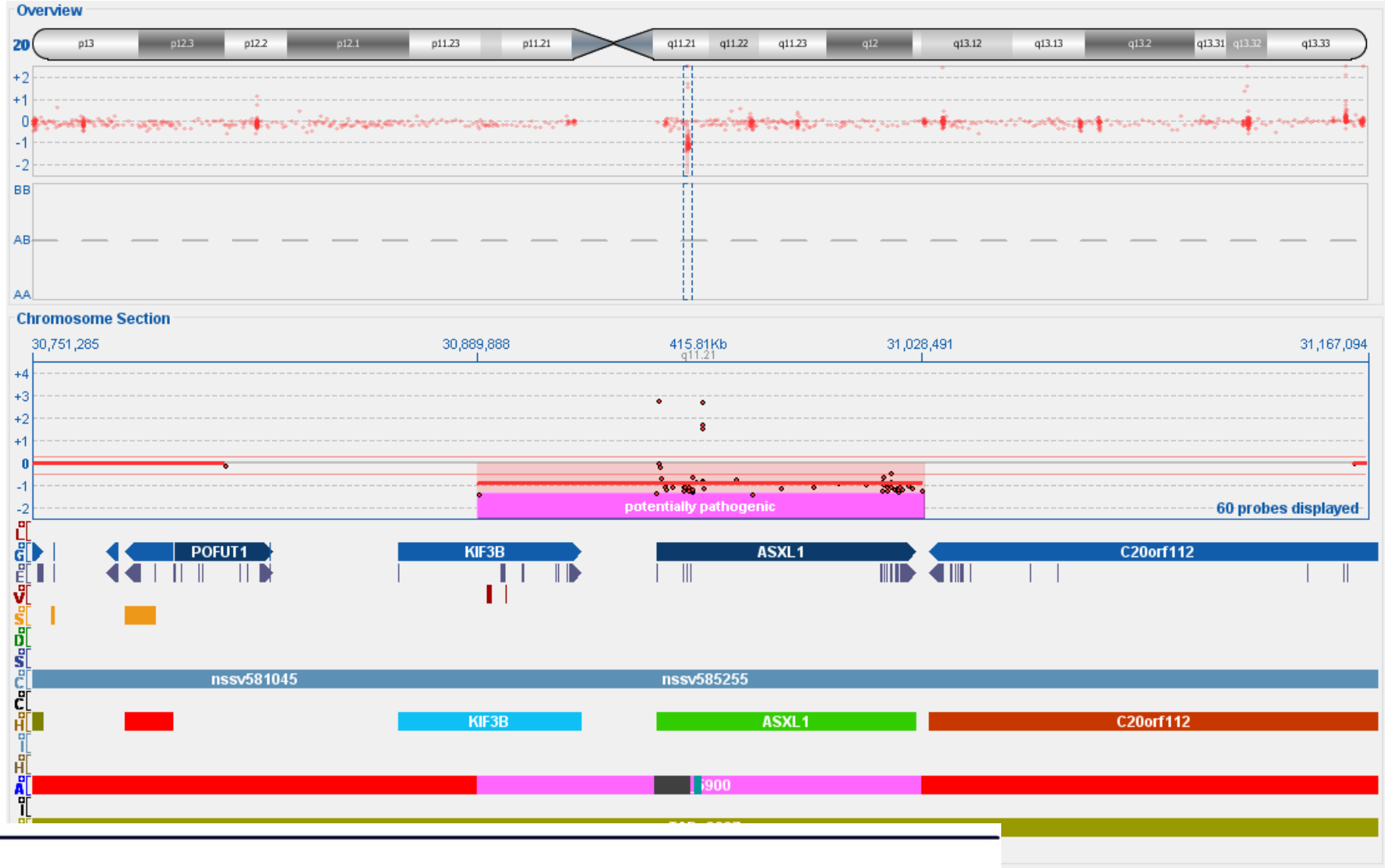
	Variation *Location*	Gene(s)	Protein change	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession
1.	GRCh38/hg38 11p15.1-13(chr11:20079474-34463996)x1 *GRCh37*: Chr11:20101020-34485543 *GRCh38*: Chr11:20079474-34463996	ABTB2, ANO3, ANO3-AS1, ANO5, ARL14EP, ARL14EP-DT, BBOX1, BBOX1-AS1, BDNF, BDNF-AS, C11orf91, CAPRIN1, CAT, CCDC179, CCDC34, CCDC73, CD59, CSTF3, CSTF3-DT, DBX1, DCDC1, DEPDC7, DNAJC24, EIF3M, ELP4, FANCF, FBXO3, FBXO3-DT ...more		See cases	Pathogenic (Aug 30, 2010)	no assertion criteria provided	VCV000154432
2.	GRCh38/hg38 11p14.3-12(chr11:22550115-38199159)x1 *GRCh37*: Chr11:22571661-38220709 *GRCh38*: Chr11:22550115-38199159	BDNF, MPPED2, CAT, CD44, CD59, CSTF3, ELF5, FANCF, FSHB, GAS2, KCNA4, LMO2, CAPRIN1, **PAX6**, RAG1, RAG2, RCN1, SLC1A2, TRAF6, WT1, PDHX, BBOX1, HIPK3, EIF3M, FJX1, KIAA1549L, ABTB2, PAMR1, FBXO3, EHF, ELP4, COMMD9, APIP ...more		See cases	Pathogenic (Aug 12, 2011)	criteria provided, single submitter	VCV000058858
3.	GRCh38/hg38 11p14.2-13(chr11:26368962-35252976)x1 *GRCh37*: Chr11:26390509-35274523 *GRCh38*: Chr11:26368962-35252976	BDNF, MPPED2, CAT, CD44, CD59, CSTF3, ELF5, FSHB, KCNA4, LMO2, CAPRIN1, **PAX6**, RCN1, SLC1A2, WT1, PDHX, BBOX1, HIPK3, EIF3M, KIAA1549L, ABTB2, FBXO3, EHF, ELP4, APIP, WT1-AS, NAT10, LIN7C, TCP11L1, LGR4, ANO3, PRRG4, OSER1 ...more		See cases	Pathogenic (Nov 30, 2010)	no assertion criteria provided	VCV000145969
4.	GRCh38/hg38 11p13(chr11:31299323-31790071)x1 *GRCh37*: Chr11:31320870-31811619 *GRCh38*: Chr11:31299323-31790071	DCDC1, DNAJC24, ELP4, IMMP1L, LOC105980003, LOC105980005, LOC105980073, **PAX6**, PAX6DRR, **PAX6** HS8		See cases	Pathogenic (Jan 27, 2011)	no assertion criteria provided	VCV000146313
5.	GRCh38/hg38 11p13(chr11:31528093-31807593)x1 *GRCh37*: Chr11:31549640-31829141 *GRCh38*: Chr11:31528093-31807593	**PAX6**, ELP4, PAX6DRR, LOC105980003, LOC105980005, LOC105980073, LOC106007485, LOC106007493		See cases	Pathogenic (Aug 12, 2011)	criteria provided, single submitter	VCV000058886
6.	NC_000011.10:g.(?_31664397)_(31794829_?)del *GRCh37*: Chr11:31685945-31816377 *GRCh38*: Chr11:31664397-31794829	ELP4, LOC105980003, LOC105980073, **PAX6**, PAX6DRR		Aniridia 1, Irido-corneo-trabecular dysgenesis	Pathogenic (Jan 8, 2019)	criteria provided, single submitter	VCV000584225
7.	NC_000011.8:g.31698271_31794414del96144 *GRCh37*: Chr11:31741695-31837838 *GRCh38*: Chr11:31720147-31816290	LOC105980003, ELP4, LOC106007485, LOC106014249, **PAX6**, LOC106007493		Congenital aniridia	Pathogenic	no assertion criteria provided	VCV000267194

SIDEBAR

	Variation *Location*	Gene(s)	Protein change	Condition(s)	Clinical significance (Last reviewed)	Review status	Accession
☐ 27.	NM_000280.4(**PAX6**):c.1169del (p.Gly390fs) *GRCh37:* Chr11:31812272 *GRCh38:* Chr11:31790724	**PAX6**	G471fs, G189fs, G254fs, G323fs, G337fs, G390fs, G404fs, G415fs, G429fs	Aniridia 1	Pathogenic (Aug 15, 2019)	criteria provided, single submitter	VCV000800476
☐ 28.	NM_000280.4(**PAX6**):c.1160_1163del (p.Gly387fs) *GRCh37:* Chr11:31812278-31812281 *GRCh38:* Chr11:31790730-31790733	**PAX6**	G412fs, G334fs, G387fs, G186fs, G401fs, G251fs, G320fs, G426fs, G468fs	Aniridia 1	Pathogenic (Aug 15, 2019)	no assertion criteria provided	VCV000800475
☐ 29.	NM_000280.4(**PAX6**):c.1077del (p.Thr360fs) *GRCh37:* Chr11:31812364 *GRCh38:* Chr11:31790816	**PAX6**	T441fs, T159fs, T224fs, T374fs, T385fs, T293fs, T307fs, T360fs, T399fs	Congenital cataract, Aniridia 1	Pathogenic (Aug 15, 2019)	criteria provided, single submitter	VCV000217328
☐ 30.	NM_000280.4(**PAX6**):c.1061_1070del (p.Tyr354fs) *GRCh37:* Chr11:31812371-31812380 *GRCh38:* Chr11:31790823-31790832	**PAX6**	Y218fs, Y287fs, Y354fs, Y368fs, Y301fs, Y379fs,	Aniridia 1	Pathogenic (Aug 15, 2019)	no assertion criteria provided	VCV000800473

Wskazanie: agenezja ciała modzelowatego u płodu

Wynik badania: arr[hg19] 20q11.21(30,889,888-31,028,491)x1 352.06Kb



Search OMIM... Options Display: Highlights

*612990
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* 612990

ASXL TRANSCRIPTIONAL REGULATOR 1; ASXL1

Alternative titles; symbols

ADDITIONAL SEX COMBS-LIKE 1
KIAA0978

HGNC Approved Gene Symbol: ASXL1

Cytogenetic location: 20q11.21 ***Genomic coordinates (GRCh38): 20:32,358,061-32,439,318*** (from NCBI)

Gene-Phenotype Relationships

View clinical synopses as a table

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
20q11.21	Bohring-Opitz syndrome	605039	AD	3
	Myelodysplastic syndrome, somatic	614286		3

PheneGene Graphics

TEXT

Description

ASXL1 is a human homolog of the Drosophila asx gene. Drosophila asx is an enhancer of trithorax

#605039
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MIM Entry

SKIN, NAILS, & HAIR

Skin

- Sacral dimple
- Nevi flammei (philtrum, nape of neck, forehead)

Hair

- Long hair
- Thick hair
- Hirsutism
- Low frontal hairline

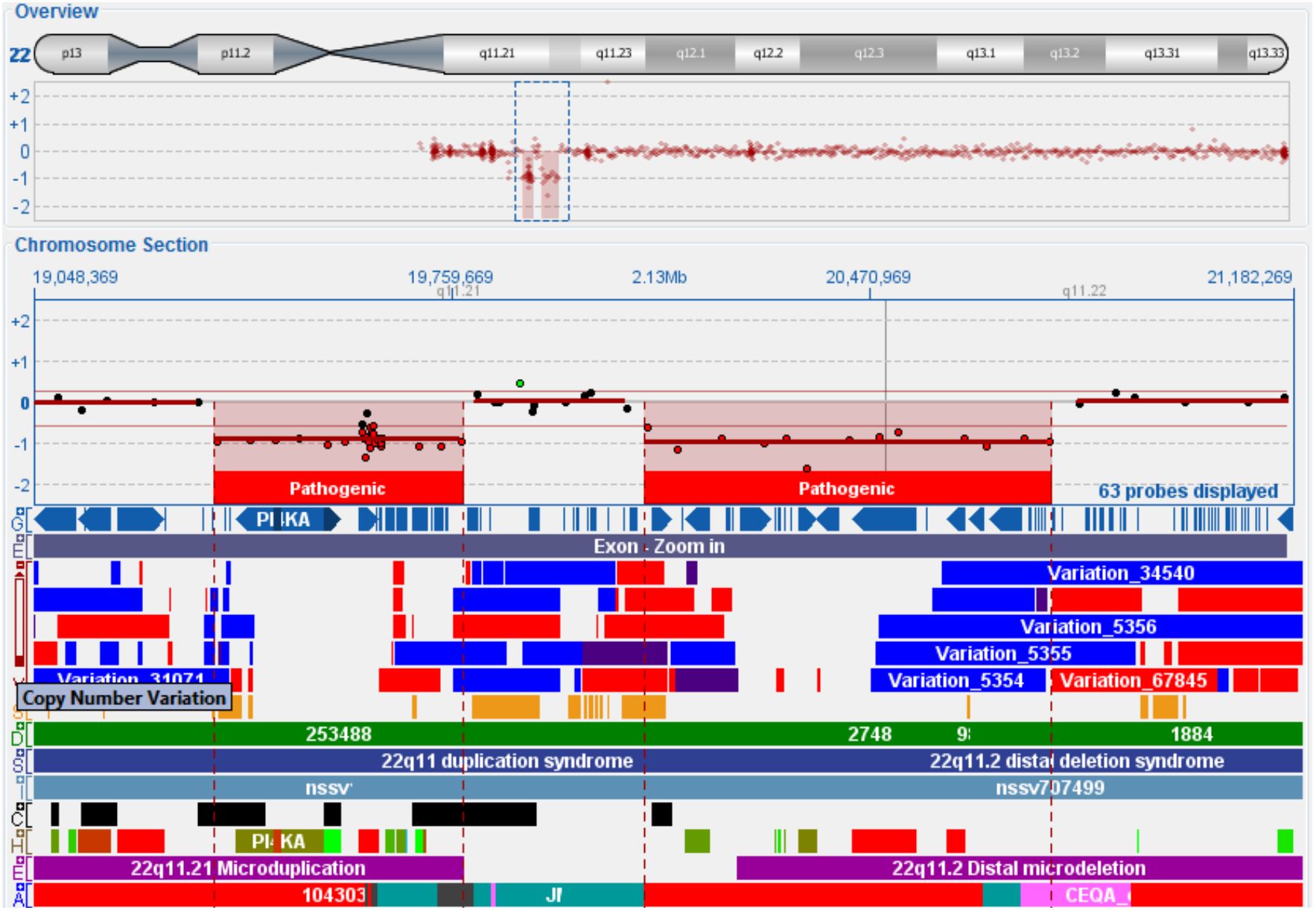
NEUROLOGIC

Central Nervous System

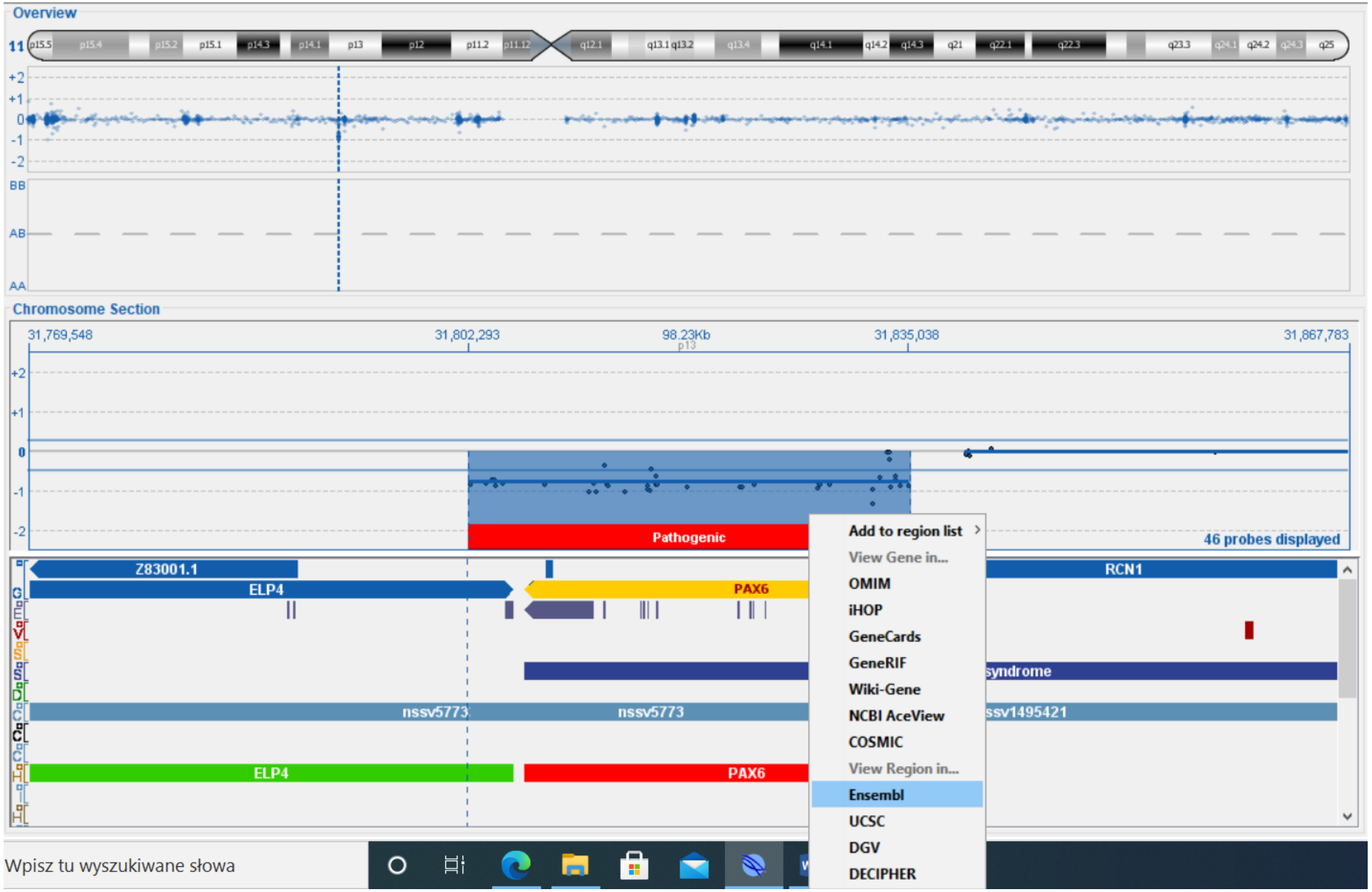
- Developmental delay
- Mental retardation, profound
- Seizures
- Hypotonia
- Agenesis of the corpus callosum
- Hypoplastic corpus callosum
- Focal nodular heterotopia
- Small brainstem
- Dandy-Walker malformation

Peripheral Nervous System

- Delayed myelination



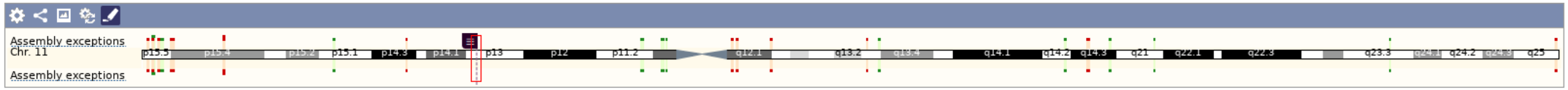
Informacje o regionie


Overview
11
p15.5 p15.4 p15.2 p15.1 p14.3 p14.1 p13 p12 p11.2 p11.12 q12.1 q13.1 q13.2 q13.4 q14.1 q14.2 q14.3 q21 q22.1 q22.3 q23.3 q24.1 q24.2 q24.3 q25
+2
+1
0
-1
-2
BB
AB
AA
Chromosome Section
31,769,548
31,802,293
98.23Kb
p13
31,835,038
31,867,783
Pathogenic
46 probes displayed
Z83001.1
ELP4
PAX6
RCN1
syndrome
nssv5773
nssv5773
ssv1495421
ELP4
PAX6
Add to region list
View Gene in...
OMIM
iHOP
GeneCards
GeneRIF
Wiki-Gene
NCBI AceView
COSMIC
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Wpisz tu wyszukiwane słowa

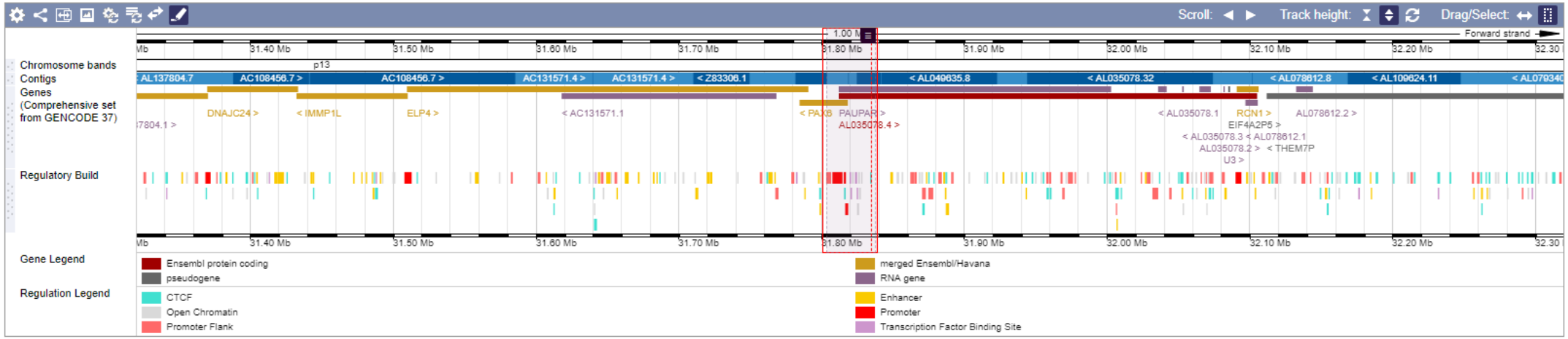
Location-based displays
- Whole genome
- Chromosome summary
- Region overview
- Region in detail
- Comparative Genomics
  - Synteny
  - Alignments (image)
  - Alignments (text)
  - Region Comparison
- Genetic Variation
  - Variant table
  - Resequencing
  - Linkage Data
- Markers
- Other genome browsers
  - UCSC
  - NCBI
  - Ensembl GRCh37

Configure this page
Custom tracks
Export data
Share this page
Bookmark this page

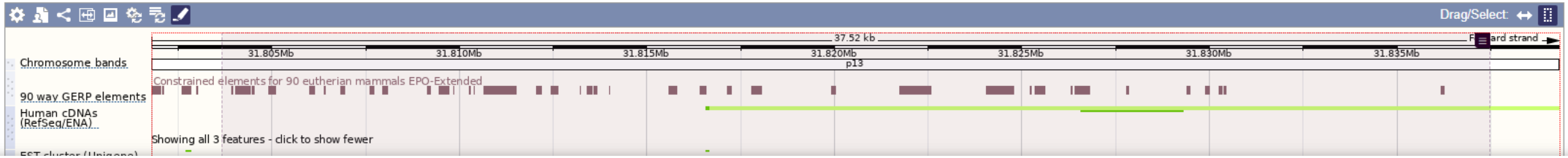
Chromosome 11: 31,801,785-31,839,308



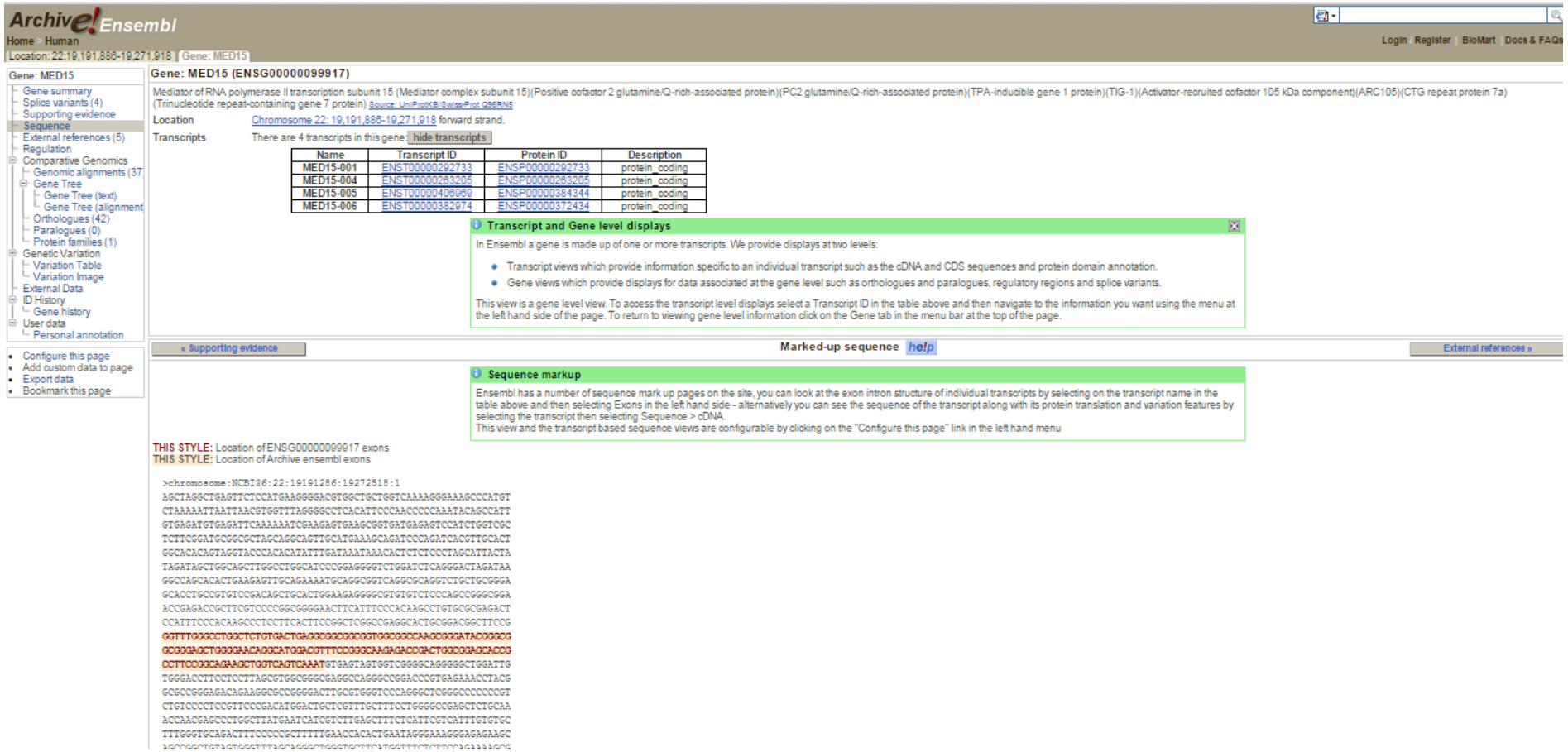
Region in detail

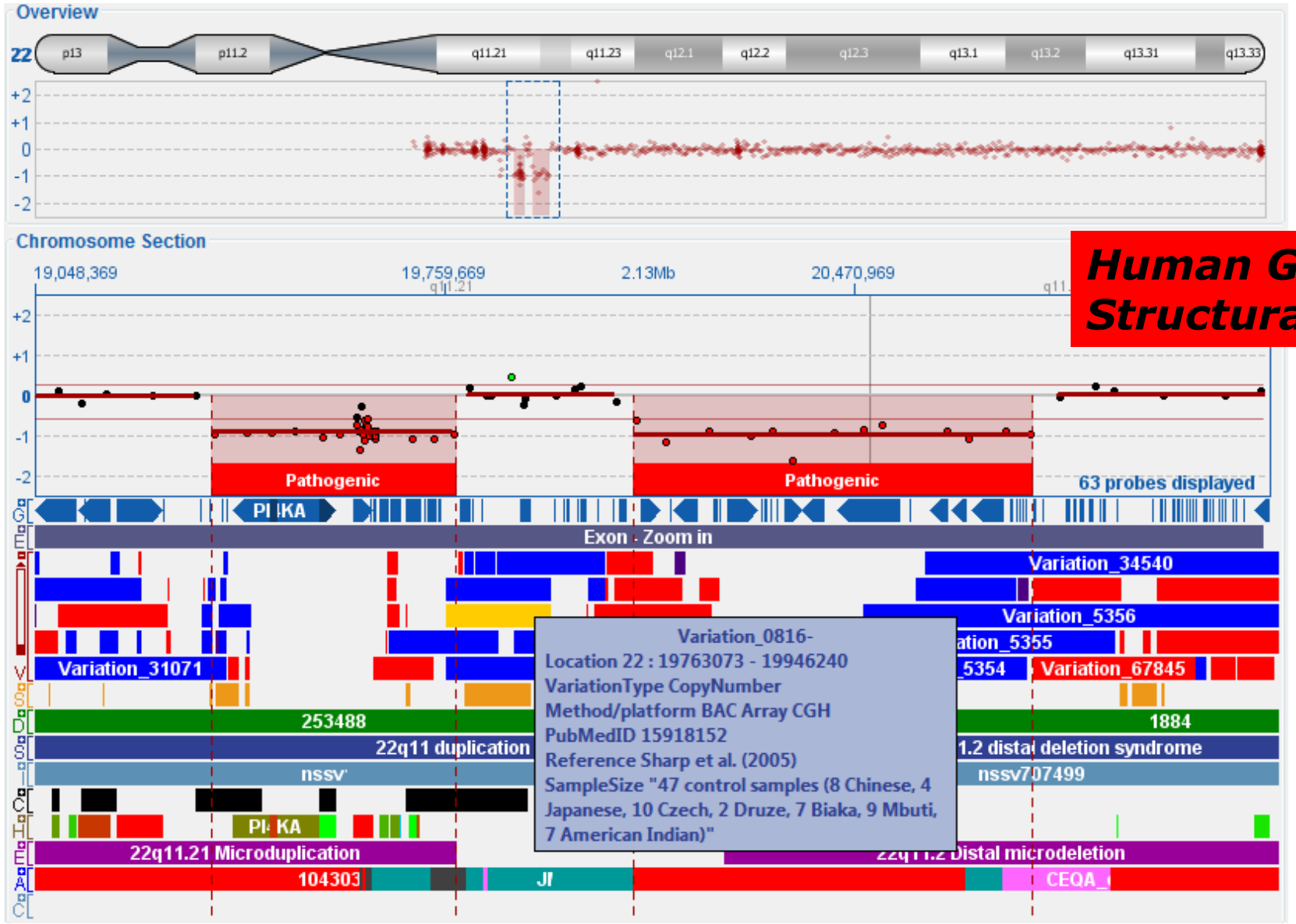


Location: 11:31801785-31839308 Go Gene: Go

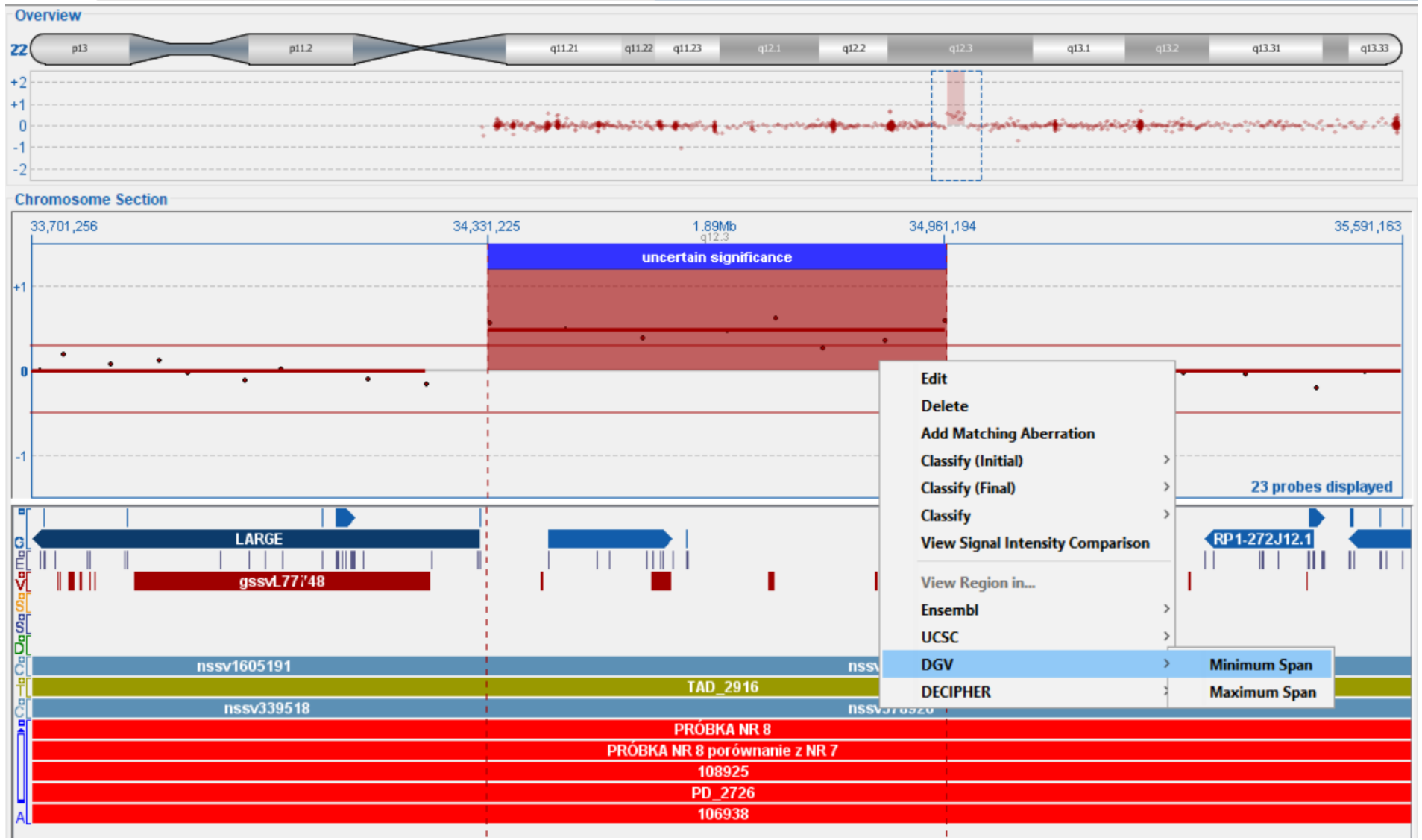


Ensembl - sekwencja genu





Human Genome Structural Variation


Overview
22
p13
p11.2
q11.21
q11.22
q11.23
q12.1
q12.2
q12.3
q13.1
q13.2
q13.31
q13.33
+2
+1
0
-1
-2
Chromosome Section
33,701,256
34,331,225
1.89Mb
q12.3
34,961,194
35,591,163
uncertain significance
+1
0
-1
23 probes displayed
LARGE
gssvL77i48
RP1-272J12.1
nssv1605191
TAD_2916
nssv339518
PRÓBKA NR 8
PRÓBKA NR 8 porównanie z NR 7
108925
PD_2726
106938
Edit
Delete
Add Matching Aberration
Classify (Initial)
Classify (Final)
Classify
View Signal Intensity Comparison
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Minimum Span
Maximum Span

Niezabezpieczona | dgv.tcag.ca/gb2/gbrowse/dgv2_hg19/?name=chr22:34331225..34961194

Database of Genomic Variants

A curated catalogue of human genomic structural variation

File Pomoc

Genomic Variants in Human Genome (Build GRCh37: Feb. 2009, hg19): Widoczne 630 kbp z sekwencji chr22, fragment od 34,331,225 do 34,961,194

Browser | Select Tracks | Custom Tracks | Preferences

Szukaj

Nazwa lub fragment:
chr22:34,331,225..34,961,194 Szukaj

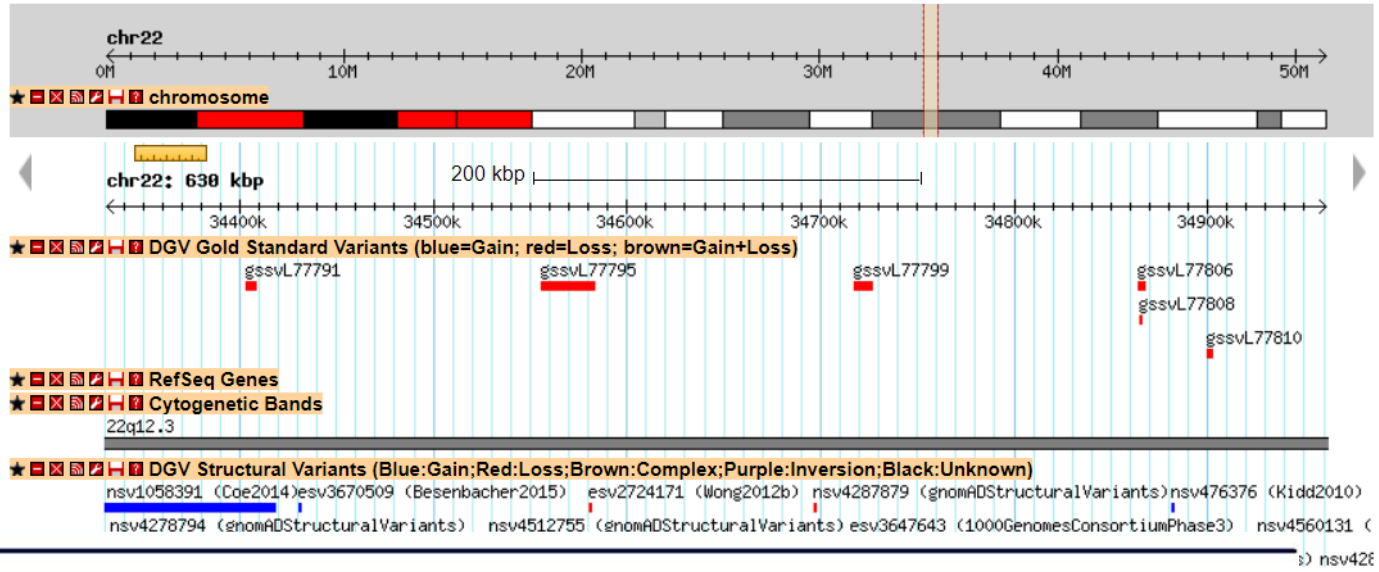
Przykłady: chr7:71890181..72690180, CFTR, AC108171.3, nsv529033.

Źródło danych
Genomic Variants in Human Genome (Build GRCh37: Feb. 2009, hg19)

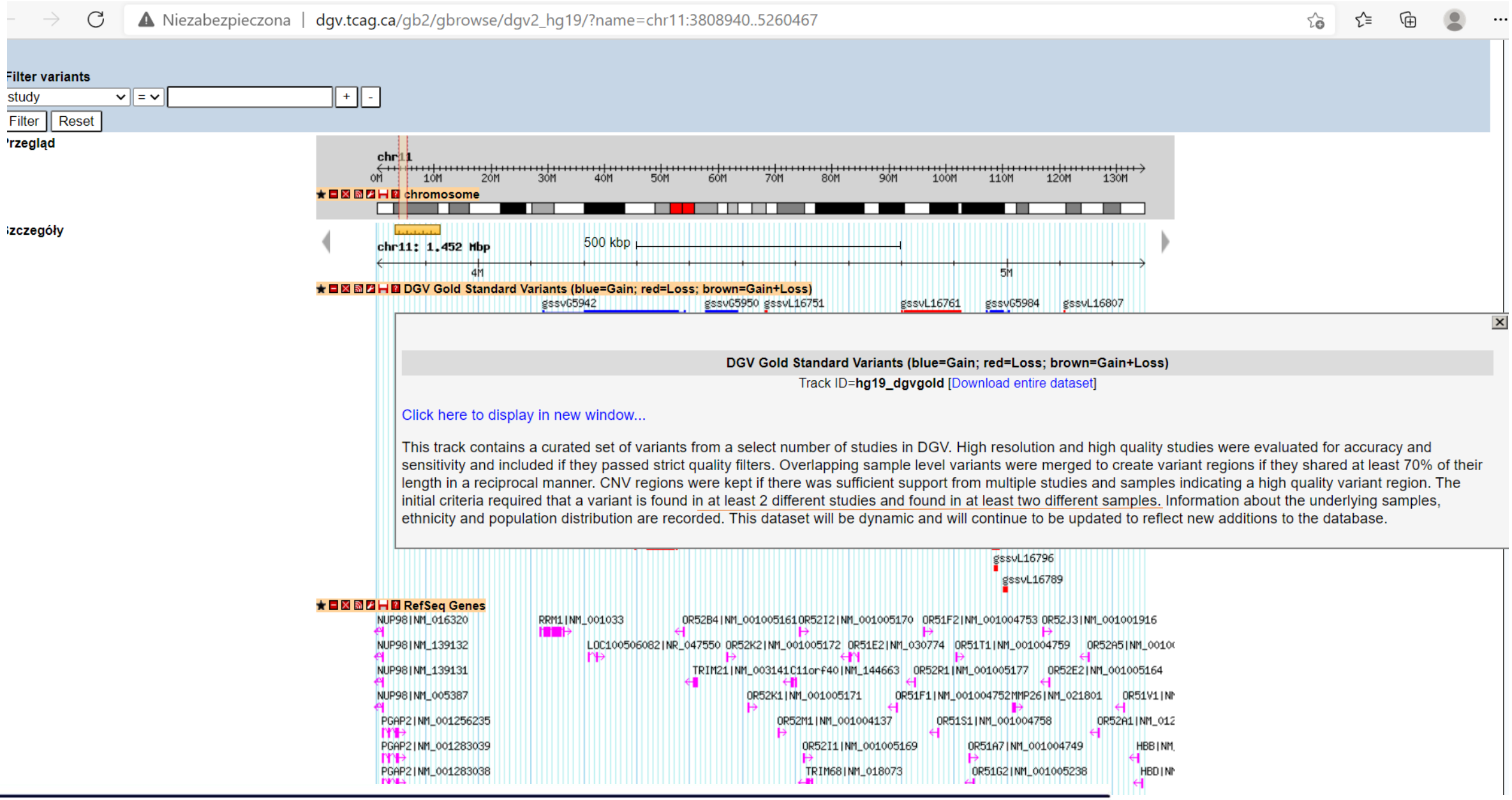
Przewijanie/powiększenie: Widoczno➔ć 630 kbp **Skieruj oś przeciwnie**

Filter variants
study = + -
Filter Reset

Przegląd



Szczegóły


Niezabezpieczona | dgv.tcag.ca/gb2/gbrowse/dgv2_hg19/?name=chr11:3808940..5260467
Filter variants
study
=
+
-
Filter
Reset
Przegląd
Szczegóły
chr11
0M 10M 20M 30M 40M 50M 60M 70M 80M 90M 100M 110M 120M 130M
chromosome
chr11: 1.452 Mbp
500 kbp
4M
5M
DGV Gold Standard Variants (blue=Gain; red=Loss; brown=Gain+Loss)
gssvG5942
gssvG5950
gssvL16751
gssvL16761
gssvG5984
gssvL16807
DGV Gold Standard Variants (blue=Gain; red=Loss; brown=Gain+Loss)
Track ID=hg19_dgvgold [Download entire dataset]
Click here to display in new window...
This track contains a curated set of variants from a select number of studies in DGV. High resolution and high quality studies were evaluated for accuracy and sensitivity and included if they passed strict quality filters. Overlapping sample level variants were merged to create variant regions if they shared at least 70% of their length in a reciprocal manner. CNV regions were kept if there was sufficient support from multiple studies and samples indicating a high quality variant region. The initial criteria required that a variant is found in at least 2 different studies and found in at least two different samples. Information about the underlying samples, ethnicity and population distribution are recorded. This dataset will be dynamic and will continue to be updated to reflect new additions to the database.
gssvL16796
gssvL16789
RefSeq Genes
NUP98|NM_016320
RRM1|NM_001033
OR52B4|NM_001005161
OR52I2|NM_001005170
OR51F2|NM_001004753
OR52J3|NM_001001916
NUP98|NM_139132
LOC100506082|NR_047550
OR52K2|NM_001005172
OR51E2|NM_030774
OR51T1|NM_001004759
OR52A5|NM_0010
NUP98|NM_139131
TRIM21|NM_003141
C11orf40|NM_144663
OR52R1|NM_001005177
OR52E2|NM_001005164
NUP98|NM_005387
OR52K1|NM_001005171
OR51F1|NM_001004752
MMP26|NM_021801
OR51V1|NM
PGAP2|NM_001256235
OR52M1|NM_001004137
OR51S1|NM_001004758
OR52A1|NM_012
PGAP2|NM_001283039
OR52I1|NM_001005169
OR51A7|NM_001004749
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PGAP2|NM_001283038
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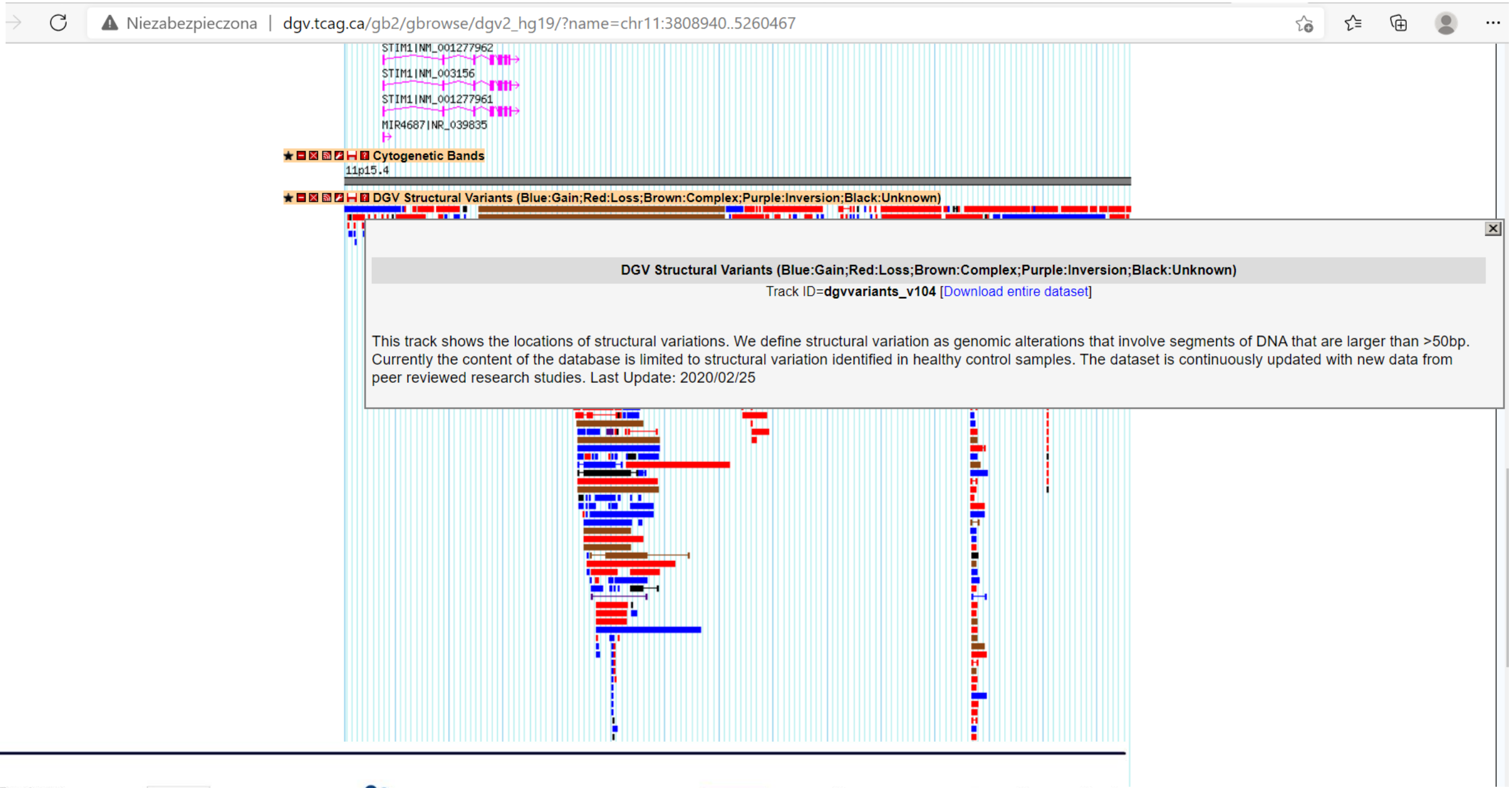
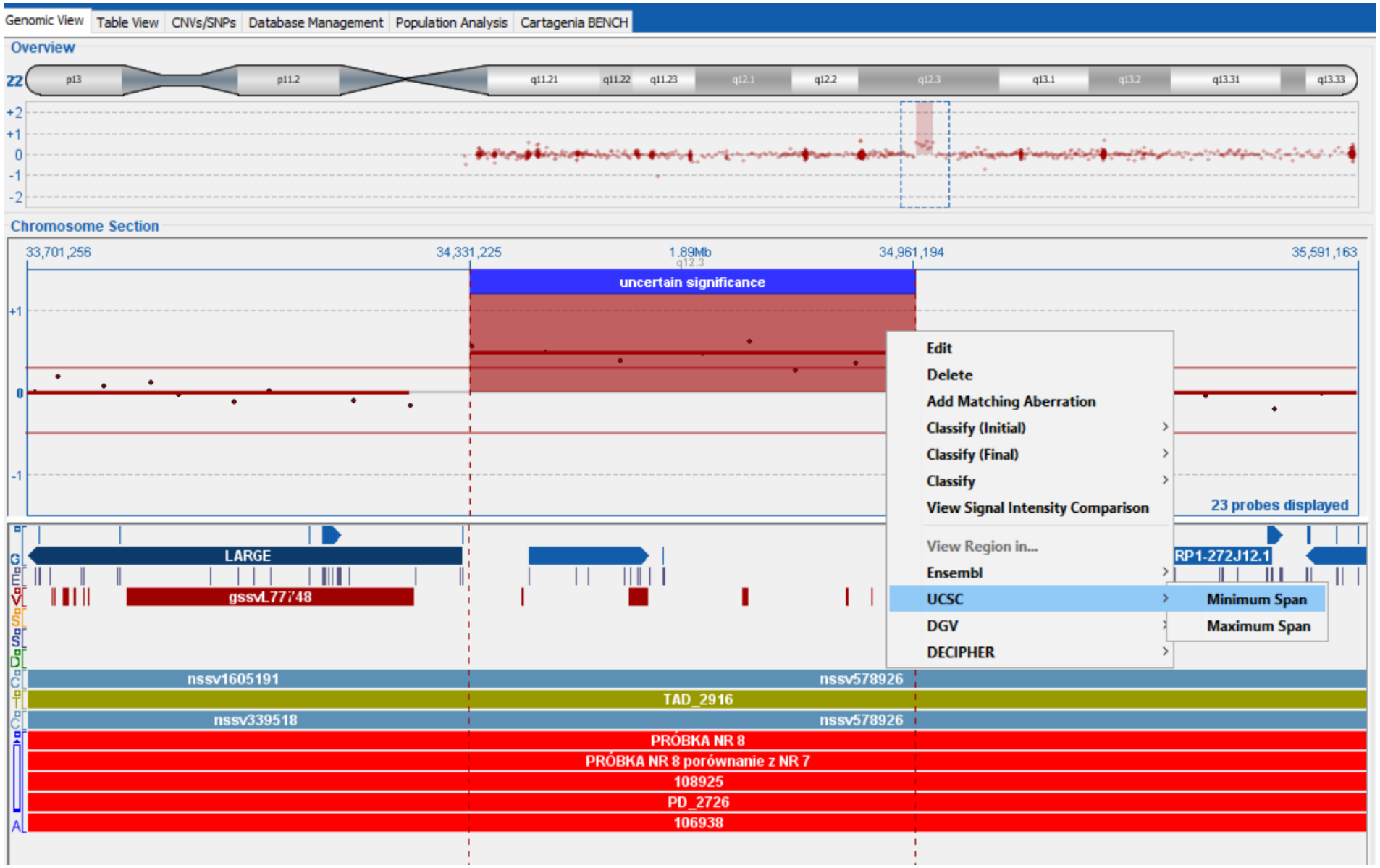
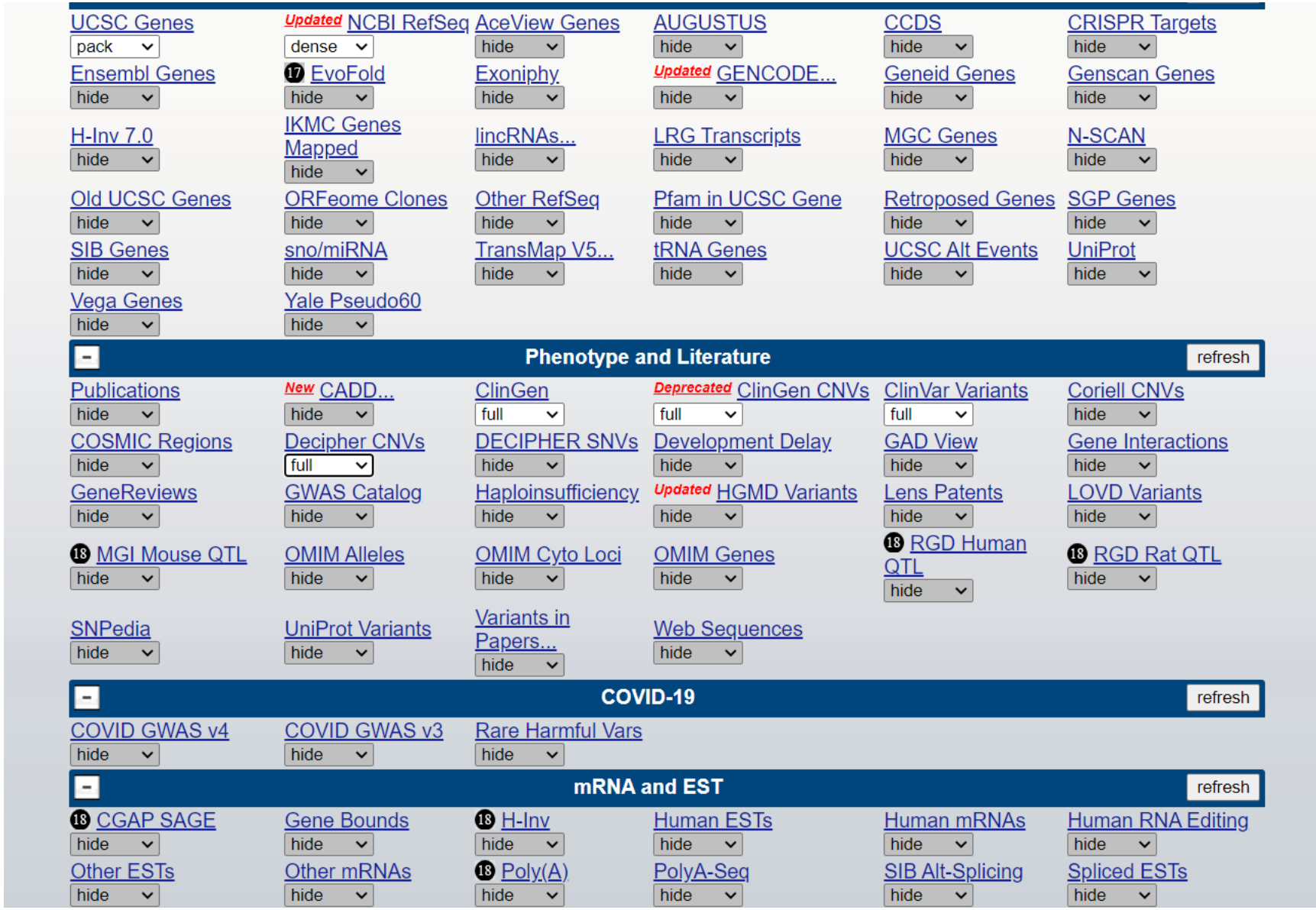
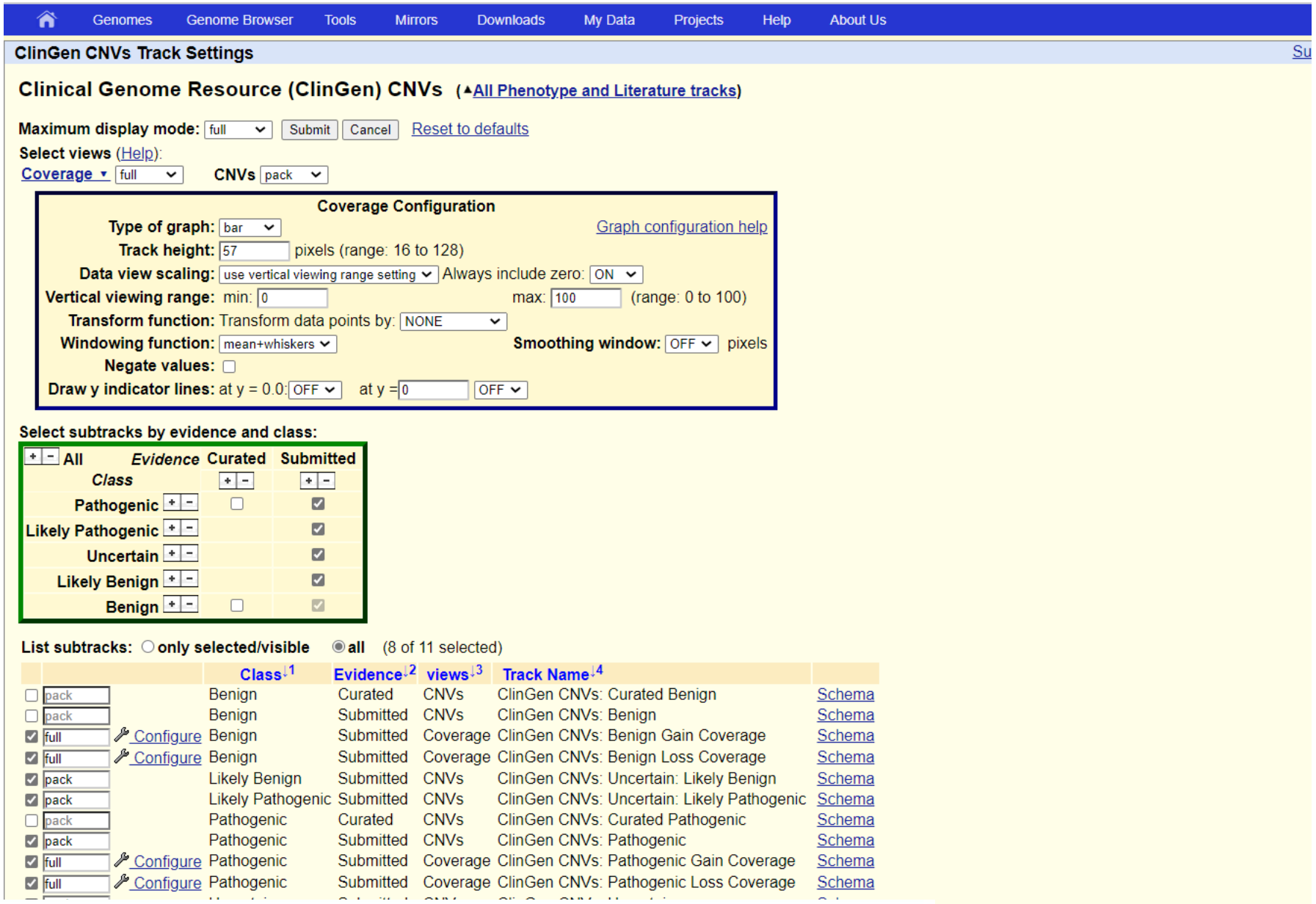
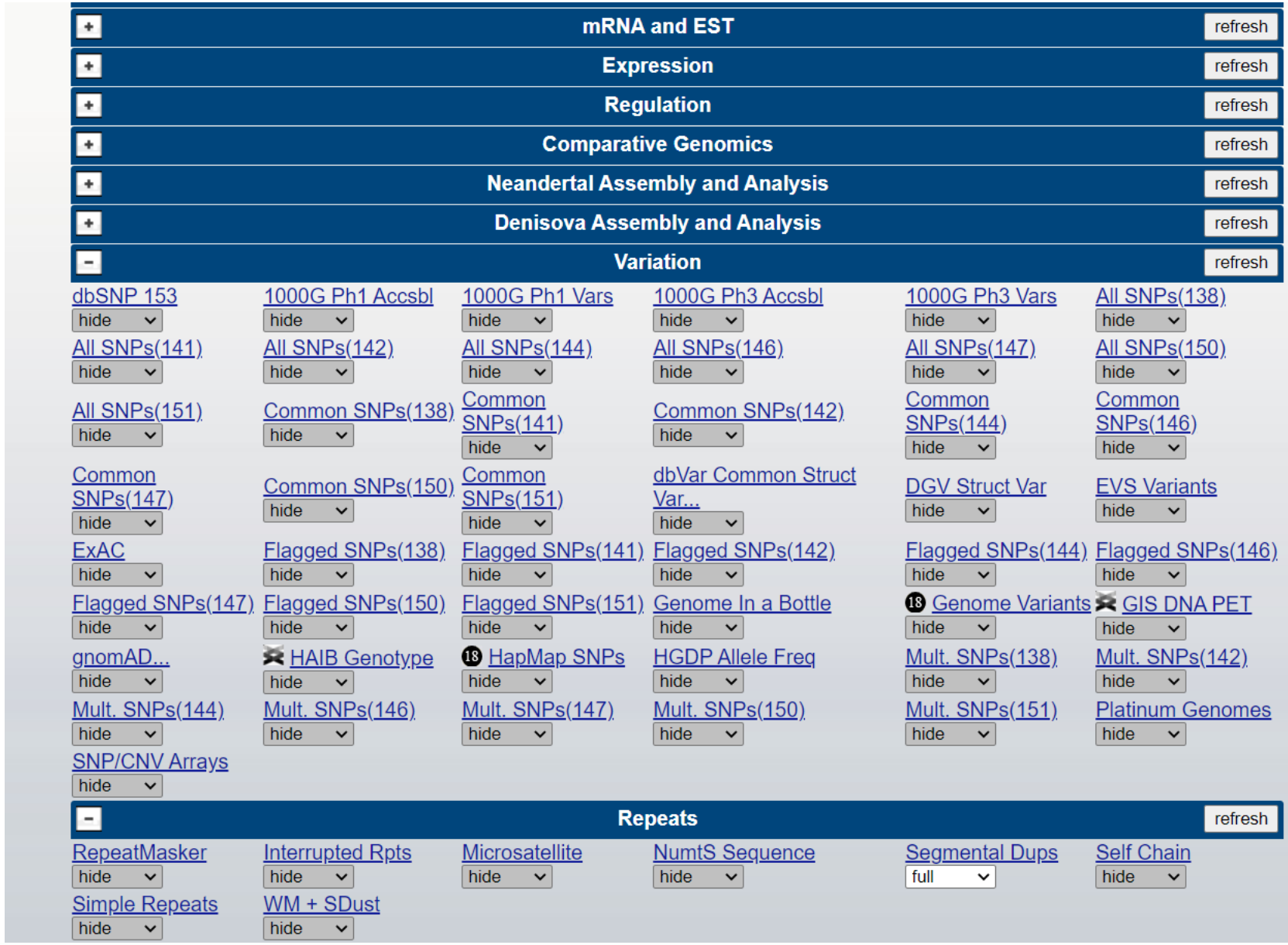
OR51G2|NM_001005238
HBD|NM

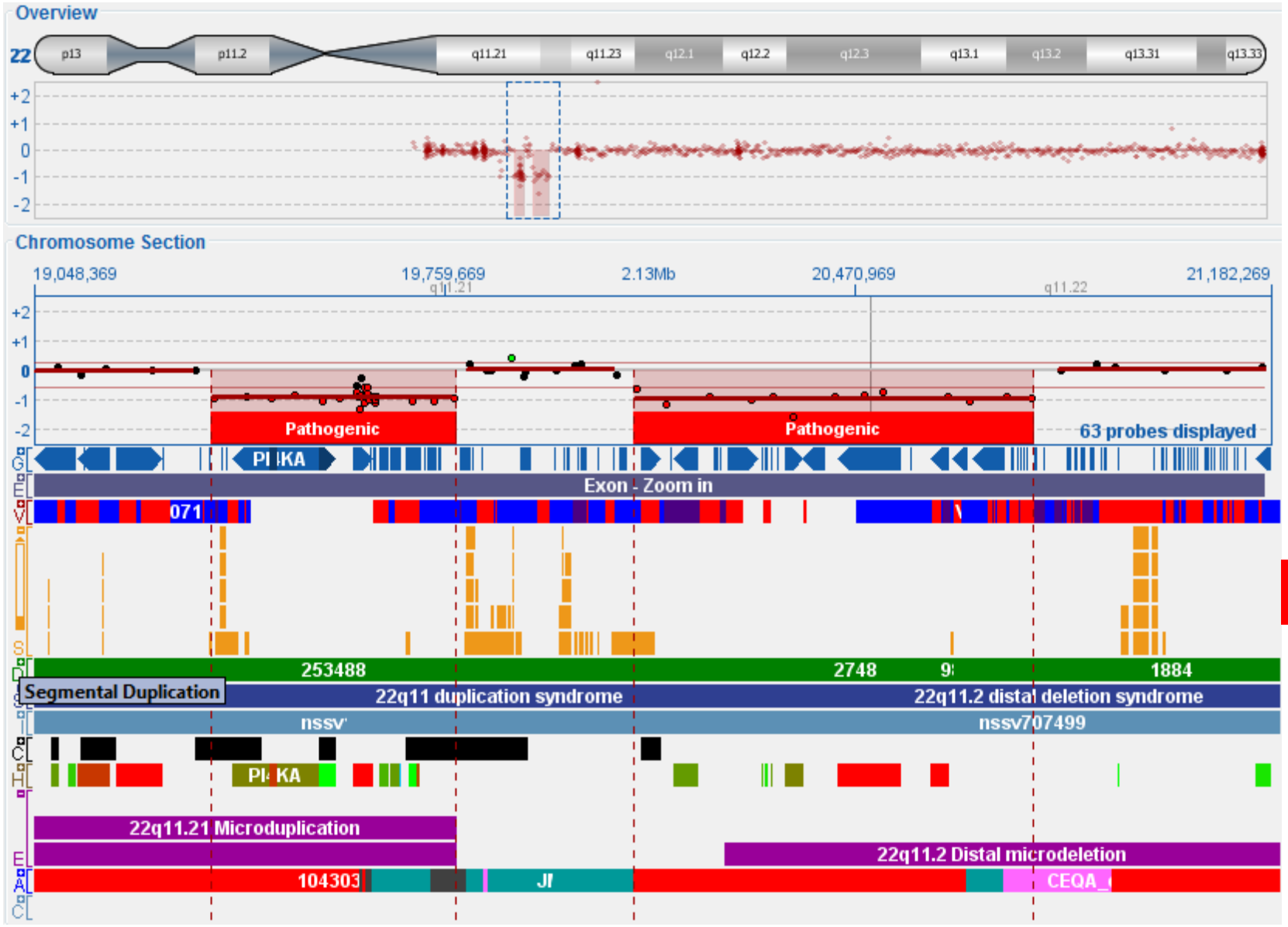

Niezabezpieczona | dgv.tcag.ca/gb2/gbrowse/dgv2_hg19/?name=chr11:3808940..5260467
STIM1|NM_001277962
STIM1|NM_003156
STIM1|NM_001277961
MIR4687|NR_039835
Cytogenetic Bands
11p15.4
DGV Structural Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown)
DGV Structural Variants (Blue:Gain;Red:Loss;Brown:Complex;Purple:Inversion;Black:Unknown)
Track ID=dgvvariants_v104 [Download entire dataset]
This track shows the locations of structural variations. We define structural variation as genomic alterations that involve segments of DNA that are larger than >50bp. Currently the content of the database is limited to structural variation identified in healthy control samples. The dataset is continuously updated with new data from peer reviewed research studies. Last Update: 2020/02/25


Genomic View
Table View
CNVs/SNPs
Database Management
Population Analysis
Cartagenia BENCH
Overview
22
p13
p11.2
q11.21
q11.22
q11.23
q12.1
q12.2
q12.3
q13.1
q13.2
q13.31
q13.33
+2
+1
0
-1
-2
Chromosome Section
33,701,256
34,331,225
1.89Mb
q12.3
34,961,194
35,591,163
uncertain significance
+1
0
-1
23 probes displayed
Edit
Delete
Add Matching Aberration
Classify (Initial)
Classify (Final)
Classify
View Signal Intensity Comparison
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Minimum Span
Maximum Span
LARGE
RP1-272J12.1
gssvL77i48
nssv1605191
nssv578926
TAD_2916
nssv339518
nssv578926
PRÓBKA NR 8
PRÓBKA NR 8 porównanie z NR 7
108925
PD_2726
106938

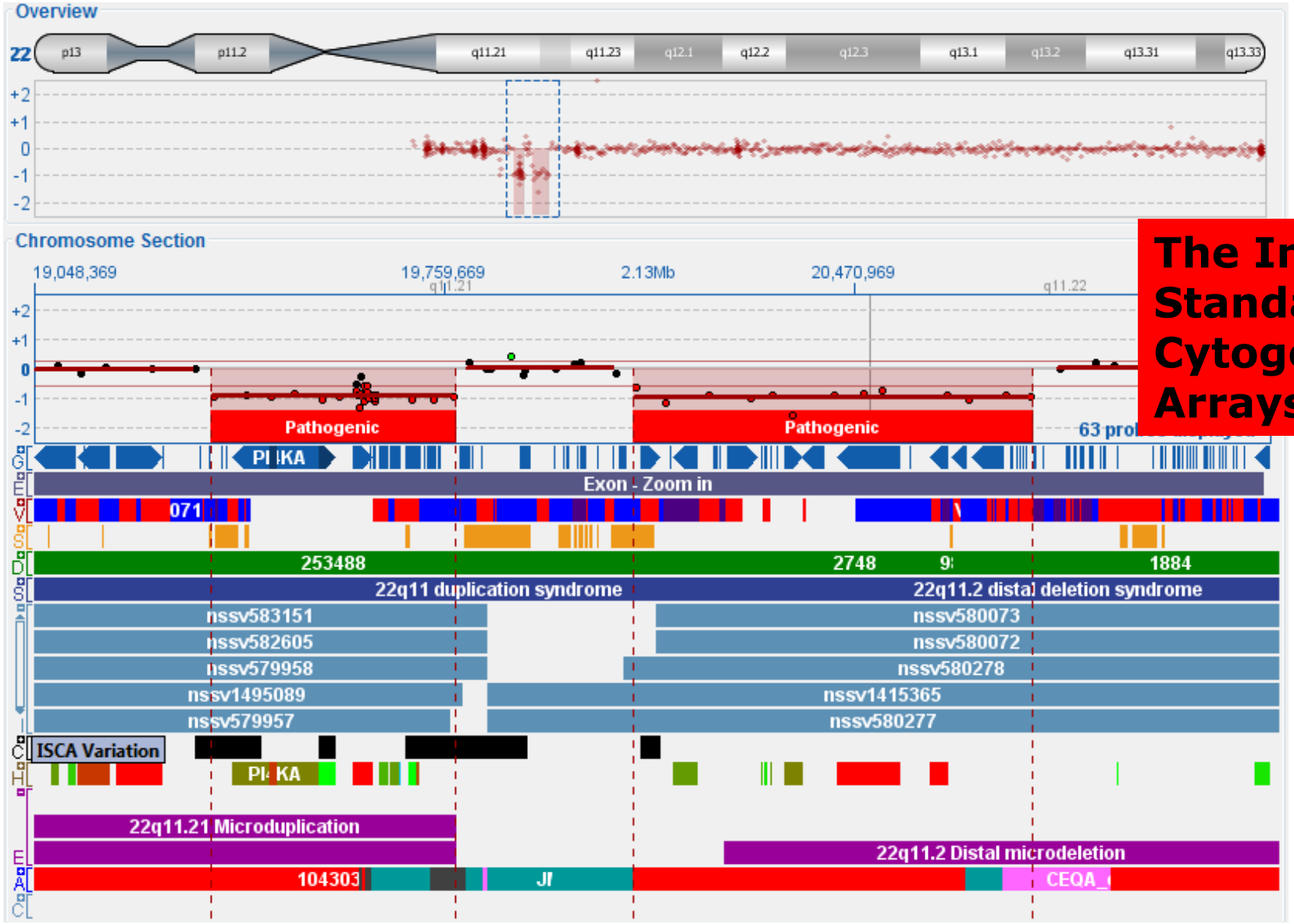

UCSC Genes
pack
Updated NCBI RefSeq
dense
AceView Genes
hide
AUGUSTUS
hide
CCDS
hide
CRISPR Targets
hide
Ensembl Genes
hide
17 EvoFold
hide
Exoniphy
hide
Updated GENCODE...
hide
Geneid Genes
hide
Genscan Genes
hide
H-Inv 7.0
hide
IKMC Genes Mapped
hide
lincRNAs...
hide
LRG Transcripts
hide
MGC Genes
hide
N-SCAN
hide
Old UCSC Genes
hide
ORFeome Clones
hide
Other RefSeq
hide
Pfam in UCSC Gene
hide
Retroposed Genes
hide
SGP Genes
hide
SIB Genes
hide
sno/miRNA
hide
TransMap V5...
hide
tRNA Genes
hide
UCSC Alt Events
hide
UniProt
hide
Vega Genes
hide
Yale Pseudo60
hide
Phenotype and Literature
refresh
Publications
hide
New CADD...
hide
ClinGen
full
Deprecated ClinGen CNVs
full
ClinVar Variants
full
Coriell CNVs
hide
COSMIC Regions
hide
Decipher CNVs
full
DECIPHER SNVs
hide
Development Delay
hide
GAD View
hide
Gene Interactions
hide
GeneReviews
hide
GWAS Catalog
hide
Haploinsufficiency
hide
Updated HGMD Variants
hide
Lens Patents
hide
LOVD Variants
hide
18 MGI Mouse QTL
hide
OMIM Alleles
hide
OMIM Cyto Loci
hide
OMIM Genes
hide
18 RGD Human QTL
hide
18 RGD Rat QTL
hide
SNPedia
hide
UniProt Variants
hide
Variants in Papers...
hide
Web Sequences
hide
COVID-19
refresh
COVID GWAS v4
hide
COVID GWAS v3
hide
Rare Harmful Vars
hide
mRNA and EST
refresh
18 CGAP SAGE
hide
Gene Bounds
hide
18 H-Inv
hide
Human ESTs
hide
Human mRNAs
hide
Human RNA Editing
hide
Other ESTs
hide
Other mRNAs
hide
18 Poly(A)
hide
PolyA-Seq
hide
SIB Alt-Splicing
hide
Spliced ESTs
hide


Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
Projects
Help
About Us
ClinGen CNVs Track Settings
Clinical Genome Resource (ClinGen) CNVs (▲All Phenotype and Literature tracks)
Maximum display mode: full Submit Cancel Reset to defaults
Select views (Help):
Coverage full CNVs pack
Coverage Configuration
Type of graph: bar
Graph configuration help
Track height: 57 pixels (range: 16 to 128)
Data view scaling: use vertical viewing range setting Always include zero: ON
Vertical viewing range: min: 0 max: 100 (range: 0 to 100)
Transform function: Transform data points by: NONE
Windowing function: mean+whiskers Smoothing window: OFF pixels
Negate values:
Draw y indicator lines: at y = 0.0: OFF at y = 0 OFF
Select subtracks by evidence and class:
All Evidence Curated Submitted
Class
Pathogenic
Likely Pathogenic
Uncertain
Likely Benign
Benign
List subtracks: only selected/visible all (8 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
full Configure Benign Submitted Coverage ClinGen CNVs: Benign Gain Coverage Schema
full 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
full Configure Pathogenic Submitted Coverage ClinGen CNVs: Pathogenic Gain Coverage Schema
full Configure Pathogenic Submitted Coverage ClinGen CNVs: Pathogenic Loss Coverage Schema


mRNA and EST refresh
Expression refresh
Regulation refresh
Comparative Genomics refresh
Neandertal Assembly and Analysis refresh
Denisova Assembly and Analysis refresh
Variation refresh
dbSNP 153 hide
1000G Ph1 Accsbl hide
1000G Ph1 Vars hide
1000G Ph3 Accsbl hide
1000G Ph3 Vars hide
All SNPs(138) hide
All SNPs(141) hide
All SNPs(142) hide
All SNPs(144) hide
All SNPs(146) hide
All SNPs(147) hide
All SNPs(150) hide
All SNPs(151) hide
Common SNPs(138) hide
Common SNPs(141) hide
Common SNPs(142) hide
Common SNPs(144) hide
Common SNPs(146) hide
Common SNPs(147) hide
Common SNPs(150) hide
Common SNPs(151) hide
dbVar Common Struct Var... hide
DGV Struct Var hide
EVS Variants hide
ExAC hide
Flagged SNPs(138) hide
Flagged SNPs(141) hide
Flagged SNPs(142) hide
Flagged SNPs(144) hide
Flagged SNPs(146) hide
Flagged SNPs(147) hide
Flagged SNPs(150) hide
Flagged SNPs(151) 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
Mult. SNPs(138) hide
Mult. SNPs(142) hide
Mult. SNPs(144) hide
Mult. SNPs(146) hide
Mult. SNPs(147) hide
Mult. SNPs(150) hide
Mult. SNPs(151) hide
Platinum Genomes hide
SNP/CNV Arrays hide
Repeats refresh
RepeatMasker hide
Interrupted Rpts hide
Microsatellite hide
NumtS Sequence hide
Segmental Dups full
Self Chain hide
Simple Repeats hide
WM + SDust hide



Low copy repeats - LCR



The International Standards for Cytogenomic Arrays ISCA

https://www.ncbi.nlm.nih.gov/projects/gap/cgi-bin/dataset.cgi?study_id=phs000205.v2.p1&phv=98564&phd=&pha=&pht=1431&phvf=&

International Standards for Cytogenomic Arrays

dbGaP Study Accession: phs000205.v2.p1

Note: This version of the study has been superseded. See most recent version

Request Access

Study version history

Study | Phenotype Datasets | Variables | Molecular Datasets | Analyses | Documents

- Browse all datasets within this study via Advanced Search
- List all datasets within this study

Dataset Name and Accession

Dataset Name: ISCA_Subject_Phenotypes
Dataset Accession: pht001431.v2.p1

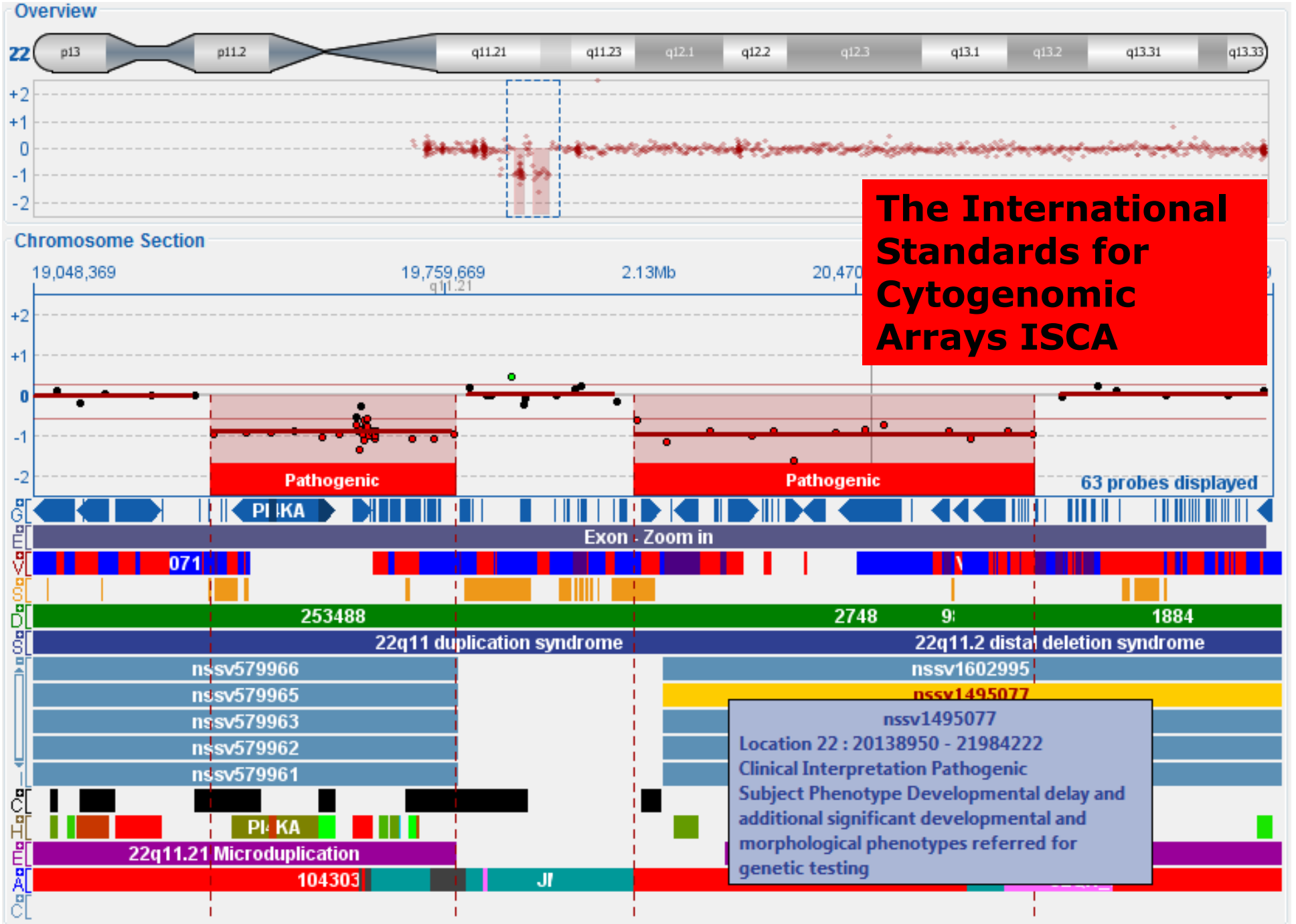
Dataset Description

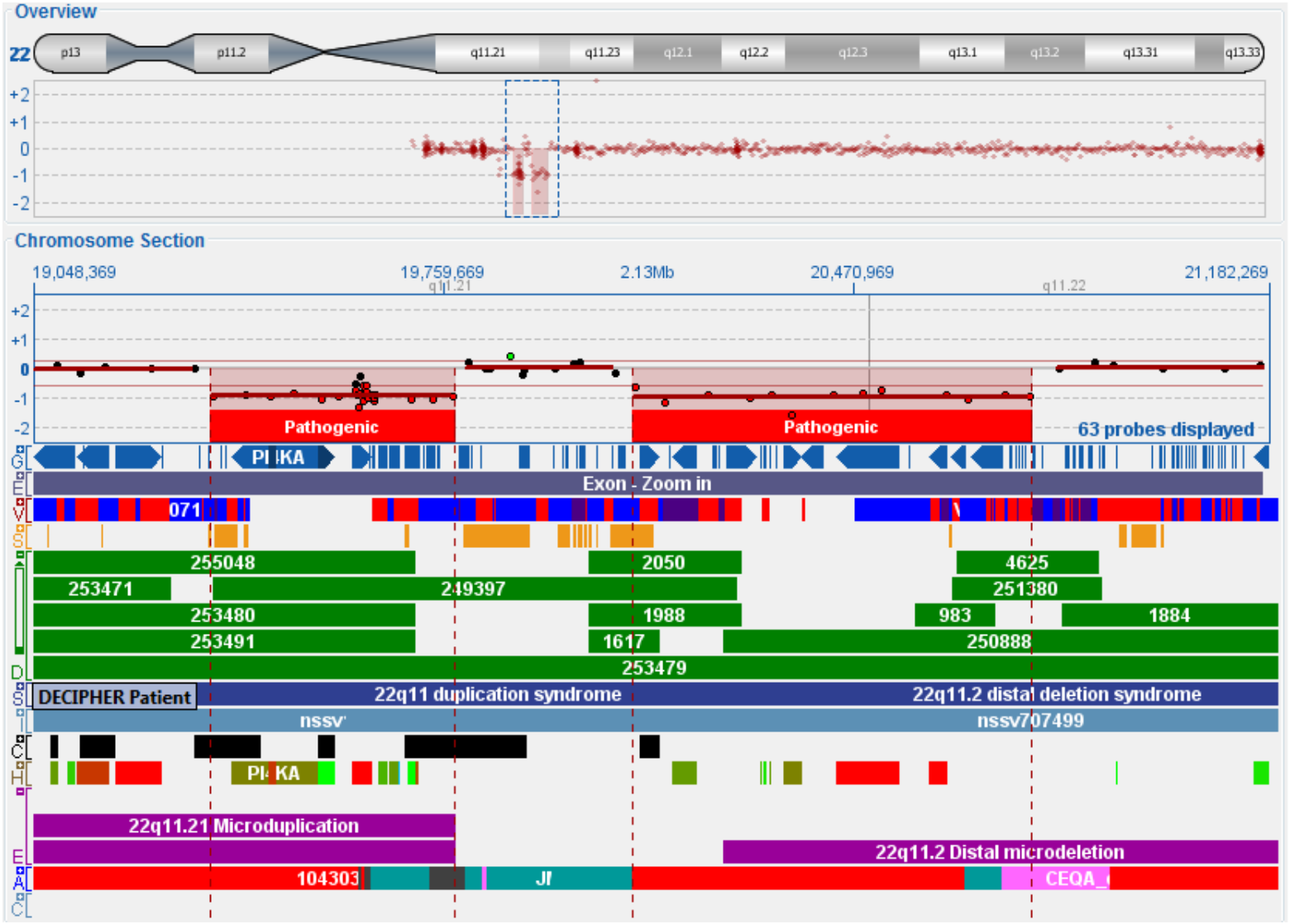
Phenotype of subjects affected with different types of genetic abnormalities

Dataset type: Simple

Dataset Summary

Download Variable Report
Download Data Dictionary


Overview
22
p13
p11.2
q11.21
q11.23
q12.1
q12.2
q12.3
q13.1
q13.2
q13.31
q13.33
+2
+1
0
-1
-2
Chromosome Section
19,048,369
19,759,669
q11.21
2.13Mb
20,470
The International Standards for Cytogenomic Arrays ISCA
Pathogenic
Pathogenic
63 probes displayed
Exon - Zoom in
253488
2748
1884
22q11 duplication syndrome
22q11.2 distal deletion syndrome
nssv579966
nssv579965
nssv579963
nssv579962
nssv579961
nssv1602995
nssv1495077
nssv1495077
Location 22 : 20138950 - 21984222
Clinical Interpretation Pathogenic
Subject Phenotype Developmental delay and additional significant developmental and morphological phenotypes referred for genetic testing
22q11.21 Microduplication
104303


Overview
22
p13
p11.2
q11.21
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q12.1
q12.2
q12.3
q13.1
q13.2
q13.31
q13.33
+2
+1
0
-1
-2
Chromosome Section
19,048,369
19,759,669
2.13Mb
20,470,969
21,182,269
q11.21
q11.22
Pathogenic
Pathogenic
63 probes displayed
Exon - Zoom in
255048
2050
4625
253471
249397
251380
253480
1988
983
1884
253491
1617
250888
253479
DECIPHER Patient
22q11 duplication syndrome
22q11.2 distal deletion syndrome
nssv
nssv707499
22q11.21 Microduplication
22q11.2 Distal microdeletion
104303

DECIPHER

https://www.deciphergenomics.org

Our URL has changed to https://www.deciphergenomics.org.
Links to https://decipher.sanger.ac.uk/ will continue to be redirected.

DECIPHER GRCh38 | About | Browse | DDD (UK) | Search DECIPHER | Help | Join | Log in

Mapping the clinical genome

Explore DECIPHER

It's free and you don't need to log in

DECIPHER is used by the clinical community to share and compare phenotypic and genotypic data. The DECIPHER database contains data from 38,423 patients who have given consent for broad data-sharing; DECIPHER also supports more limited sharing via consortia. Have a look at the numbers.

Anyone can browse publicly-available patient data on DECIPHER and request to be put in contact with the responsible clinician. Why? Because sharing benefits everyone.

Join DECIPHER

Be part of the sharing community

Projects affiliated to DECIPHER can deposit and share patients, variants, and phenotypes to invite collaboration and facilitate diagnosis. Once deposited, you can use DECIPHER to identify and prioritise potential matches, and you can request notifications as soon as new matches arrive.

As well as influencing individual patient outcomes, use of DECIPHER has contributed to over 2600 published articles since 2004. It's still free, and you are in control of what data to make public.

Already a member?

Log in to access your patient data

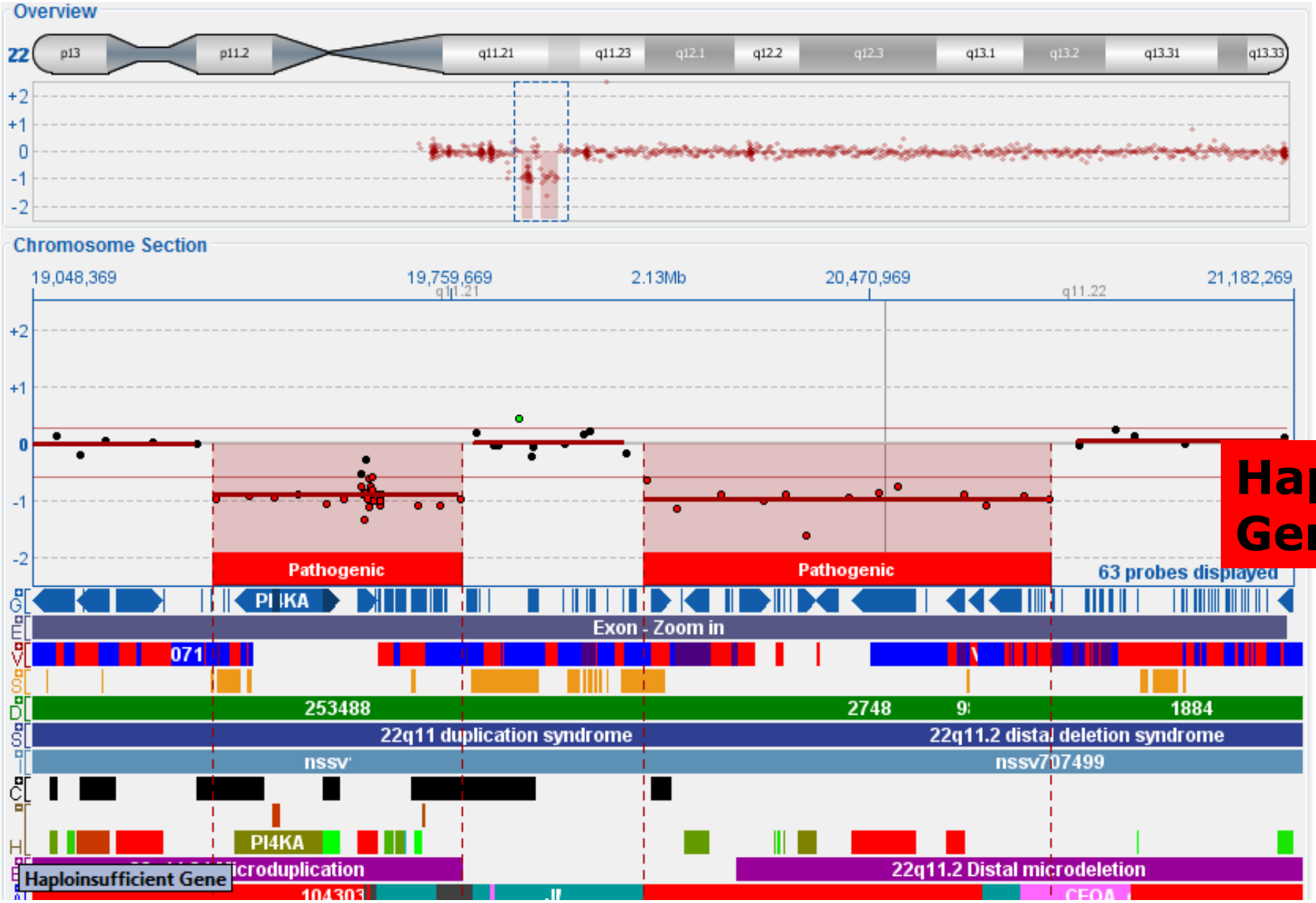
Email address

Email

Password

Password

Log in



Haploinsufficient Genes

Ocena „wrażliwości na dawkę”

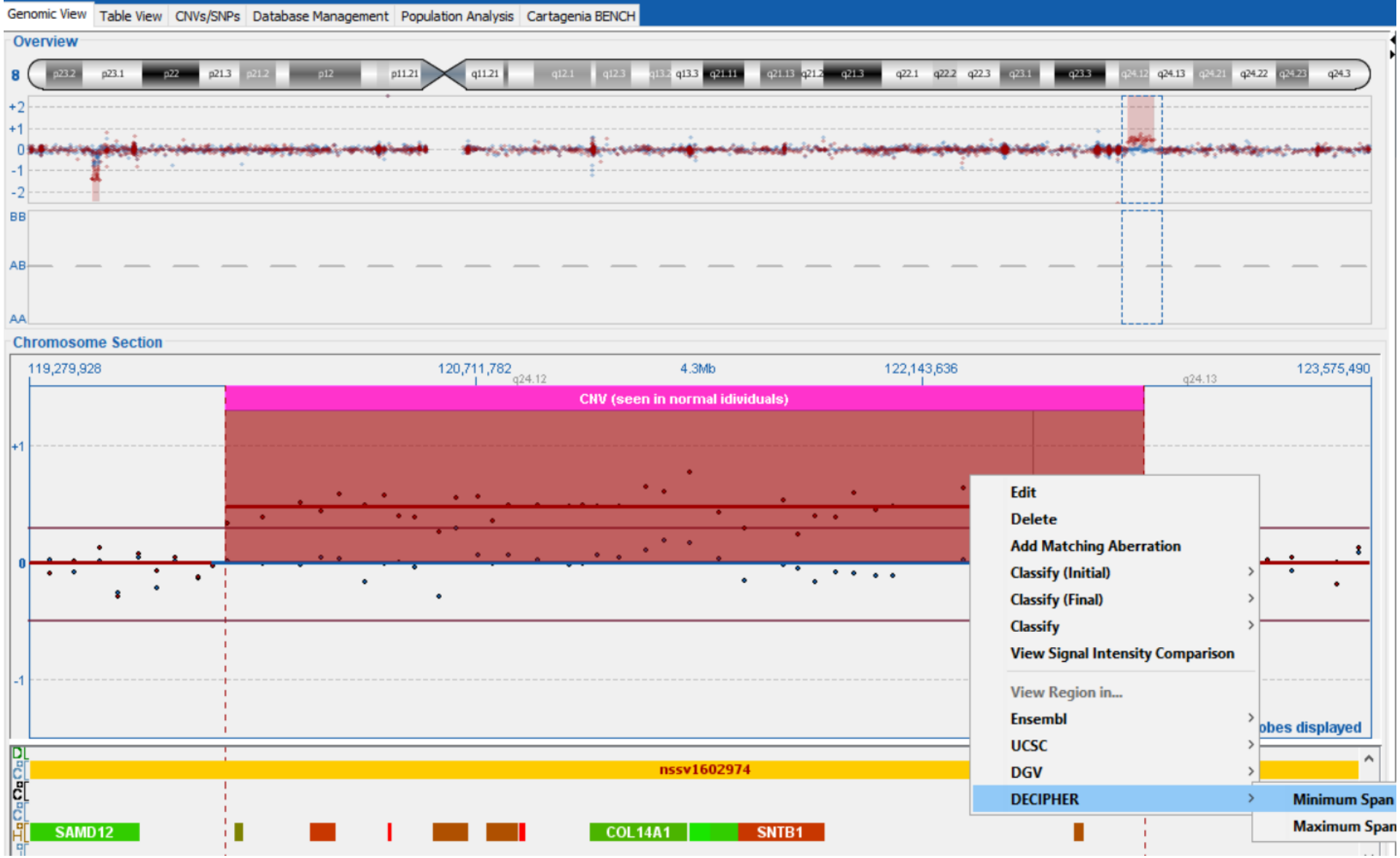
OPEN ACCESS Freely available online

PLoS GENETICS

Characterising and Predicting Haploinsufficiency in the Human Genome

Ni Huang[1], Insuk Lee[2,3], Edward M. Marcotte[2], Matthew E. Hurles[1]*

1 Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Cambridge, United Kingdom, 2 Center for Systems and Synthetic Biology, Department of Chemistry and Biochemistry, Institute for Cellular and Molecular Biology, University of Texas, Austin, Texas, United States of America, 3 Department of Biotechnology, College of Life Science and Biotechnology, Yonsei University, Seoul, South Korea


Genomic View
Table View
CNVs/SNPs
Database Management
Population Analysis
Cartagenia BENCH
Overview
Chromosome Section
119,279,928
120,711,782
4.3Mb
122,143,636
123,575,490
CNV (seen in normal idividuals)
Edit
Delete
Add Matching Aberration
Classify (Initial)
Classify (Final)
Classify
View Signal Intensity Comparison
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Minimum Span
Maximum Span
nssv1602974
SAMD12
COL14A1
SNTB1

Search results for "8:119911864-122847565" (Refine Search)

Patient variants 57 | CNV syndrome variants 0 | DDD research variants 1 | Genes 15

Results | Browser

Variants: 1 to 10 of 57

Show: All variant types Filter...

DECIPHER Patient	Sex	Location	Size	Inheritance / Genotype	Pathogenicity / Contribution	Phenotype(s)	Contact
248605	Other	8 119555793 120702839 GRCh38 Duplication	1.15 Mb	Inherited from parent with similar phenotype to child Heterozygous			
249525	Unknown	8 119629941 138371840 GRCh38 Deletion	18.74 Mb	Unknown Heterozygous		Intellectual disability	
251369	46XY	8 112882803 145068712 GRCh38	32.19 Mb	De novo Heterozygous		Intellectual disability	

DECIPHER GRCh38 About Browse DDD (UK) Search DECIPHER Help Join Log in

Search results for "8:118899625-121835325" (Refine Search)

Patient variants 68 | CNV syndrome variants 0 | DDD research variants 1 | Genes 25

Genes: 1 to 10 of 25

Show: OMIM Morbid DDG2P ClinGen Protein coding Filter...

Name / Description	Location	pLI	LOEUF	sHet	%HI	OMIM / Morbid	DDG2P	ClinGen	Links
CCN3 cellular communication network factor 3	8 119416446 119424434	0.01	0.81	-	-	OMIM	-	-	View
COL14A1 collagen type XIV alpha 1 chain	8 120059780 120373573	0.00	0.54	0.047	6.75	OMIM	-	-	View
COLEC10 collectin subfamily member 10	8 118995452 119108455	0.00	0.96	0.043	36.63	OMIM Morbid	Probable: Biallelic	-	View
CYCSP23 CYCS pseudogene 23	8 119618162 119618474	-	-	-	-	-	-	-	View
DEPTOR DEP domain containing MTOR interacting protein	8 119873717 120050918	0.00	1.43	0.010	32.17	OMIM	-	-	View

DECIPHER GRCh38 About Browse DDD (UK) Search DECIPHER Help Join Log in

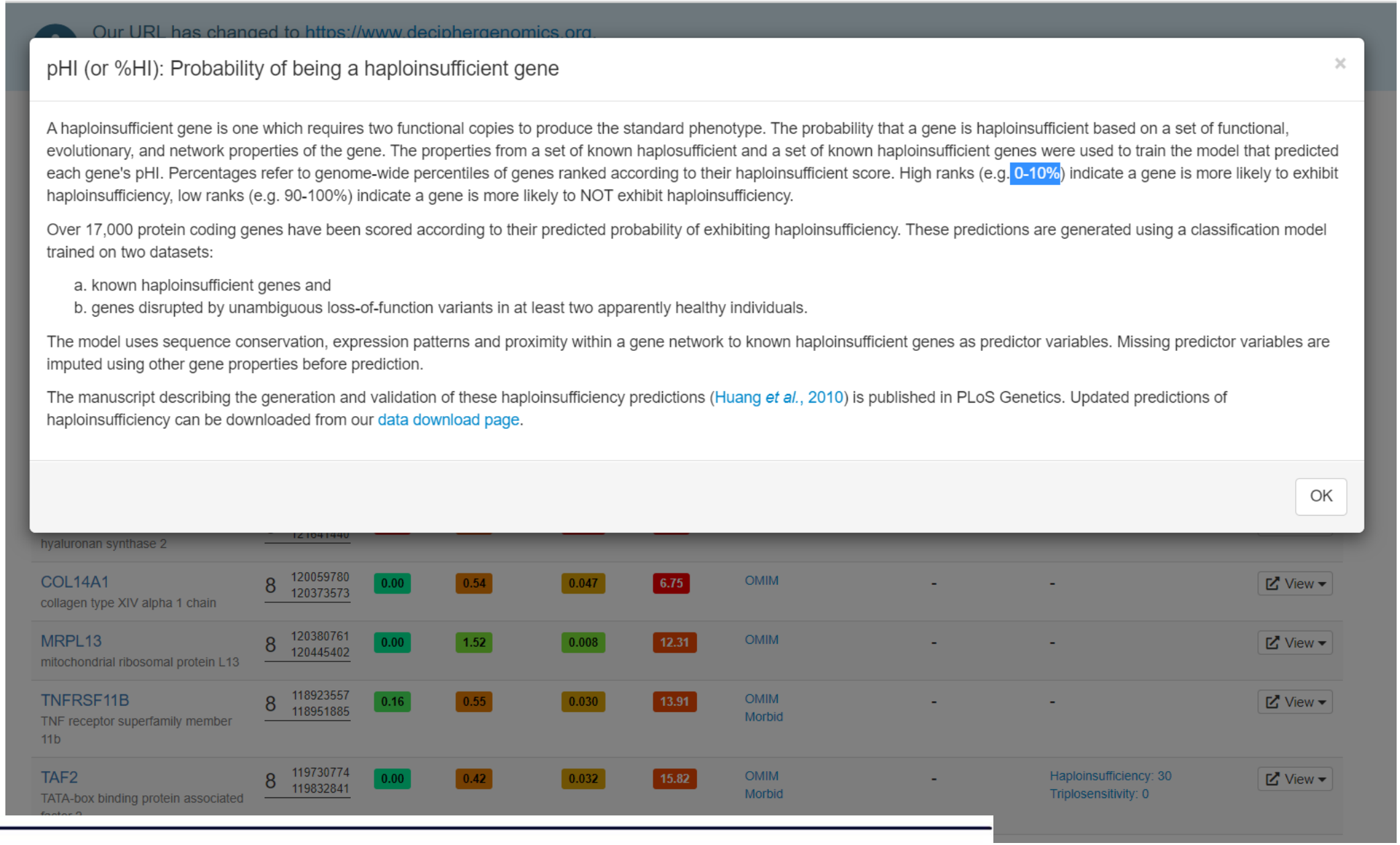
Search results for "8:118899625-121835325" (Refine Search)

Patient variants 68 | CNV syndrome variants 0 | DDD research variants 1 | Genes 25

Genes: 1 to 10 of 13 (out of 25 total)

Show: OMIM Morbid DDG2P ClinGen Protein coding Filter...

Name / Description	Location	pLI	LOEUF	sHet	%HI	OMIM / Morbid	DDG2P	ClinGen	Links
HAS2 hyaluronan synthase 2	8 121612116 121641440	0.99	0.28	0.242	4.07	OMIM	-	-	View
COL14A1 collagen type XIV alpha 1 chain	8 120059780 120373573	0.00	0.54	0.047	6.75	OMIM	-	-	View
MRPL13 mitochondrial ribosomal protein L13	8 120380761 120445402	0.00	1.52	0.008	12.31	OMIM	-	-	View
TNFRSF11B TNF receptor superfamily member 11b	8 118923557 118951885	0.16	0.55	0.030	13.91	OMIM Morbid	-	-	View
TAF2 TATA-box binding protein associated factor 2	8 119730774 119832841	0.00	0.42	0.032	15.82	OMIM Morbid	-	Haploinsufficiency: 30 Triplosensitivity: 0	View


Our URL has changed to https://www.deciphergenomics.org.
pHI (or %HI): Probability of being a haploinsufficient gene
A haploinsufficient gene is one which requires two functional copies to produce the standard phenotype. The probability that a gene is haploinsufficient based on a set of functional, evolutionary, and network properties of the gene. The properties from a set of known haplosufficient and a set of known haploinsufficient genes were used to train the model that predicted each gene's pHI. Percentages refer to genome-wide percentiles of genes ranked according to their haploinsufficient score. High ranks (e.g. 0-10%) indicate a gene is more likely to exhibit haploinsufficiency, low ranks (e.g. 90-100%) indicate a gene is more likely to NOT exhibit haploinsufficiency.
Over 17,000 protein coding genes have been scored according to their predicted probability of exhibiting haploinsufficiency. These predictions are generated using a classification model trained on two datasets:
a. known haploinsufficient genes and
b. genes disrupted by unambiguous loss-of-function variants in at least two apparently healthy individuals.
The model uses sequence conservation, expression patterns and proximity within a gene network to known haploinsufficient genes as predictor variables. Missing predictor variables are imputed using other gene properties before prediction.
The manuscript describing the generation and validation of these haploinsufficiency predictions (Huang et al., 2010) is published in PLoS Genetics. Updated predictions of haploinsufficiency can be downloaded from our data download page.
OK
hyaluronan synthase 2
COL14A1 collagen type XIV alpha 1 chain 8 120059780 120373573 0.00 0.54 0.047 6.75 OMIM - - View
MRPL13 mitochondrial ribosomal protein L13 8 120380761 120445402 0.00 1.52 0.008 12.31 OMIM - - View
TNFRSF11B TNF receptor superfamily member 11b 8 118923557 118951885 0.16 0.55 0.030 13.91 OMIM Morbid - - View
TAF2 TATA-box binding protein associated 8 119730774 119832841 0.00 0.42 0.032 15.82 OMIM Morbid - Haploinsufficiency: 30 Triplosensitivity: 0 View

Search results for "8:118899625-121835325" (Refine Search)

Patient variants 68 | CNV syndrome variants 0 | DDD research variants 1 | Genes 25

Genes: 1 to 3 of 3 (out of 25 total)

Show: ☐ OMIM ☑ Morbid ☐ DDG2P ☐ ClinGen ☑ Protein coding Filter...

Name / Description	Location	pLI	LOEUF	sHet	%HI	OMIM / Morbid	DDG2P	ClinGen	Links
TNFRSF11B TNF receptor superfamily member 11b	8 118923557 118951885	0.16	0.55	0.030	13.91	OMIM Morbid	-	-	View
TAF2 TATA-box binding protein associated factor 2	8 119730774 119832841	0.00	0.42	0.032	15.82	OMIM Morbid	-	Haploinsufficiency: 30 Triplosensitivity: 0	View
COLEC10 collectin subfamily member 10	8 118995452 119108455	0.00	0.96	0.043	36.63	OMIM Morbid	Probable: Biallelic	-	View

10 Previous 1 Next

TNFRSF11B 8:118923557-118951885

Reverse strand gene: TNF receptor superfamily member 11b

Formerly known as: **OPG**

Also known as: **TR1, OCIF, ENSG00000164761**

Function: "Acts as decoy receptor for TNFSF11/RANKL and thereby neutralizes its function in osteoclastogenesis. Inhibits the activation of osteoclasts and promotes osteoclast apoptosis in vitro. Bone homeostasis seems to depend on the local ratio between TNFSF11 and TNFRSF11B. May also play…" Show more » *Source: UniProt*

DECIPHER holds no open-access sequence variants in this gene

Overview | Matching patient variants 28 | Matching DDD research variants 0 | Phenotypes | Phenotype browser | Transcripts 3 | Browser

Clinical | Protein / Genomic

Gene/disease association

OMIM
602643

Morbid
Paget disease of bone 5, juvenile-onset (Autosomal recessive)

DDG2P
-

Gene disorders

ClinGen gene/disease
-

ClinGen DS
-

Predictive scores

pLI	0.16
%HI	13.91
LOEUF	0.55
sHet	0.030

Search databases

- PubMed
- Gene Tests
- Genomics England PanelApp
- LSDB
- Entries in DECIPHER for this gene

ClinGen Dosage Sensitivity Map

The Clinical Genome Resource (ClinGen) consortium is curating genes and regions of the genome to assess whether there is evidence to support that these genes/regions are dosage sensitive and should be targeted on a cytogenomic array.

All data are shown in GRCh37 and GRCh38 coordinates.

New! The ClinGen Dosage Sensitivity curations and downloads that are available at this site are now also available at www.clinicalgenome.org. Click on the button to access Dosage Sensitivity in the context of ClinGen's other curated information, including Gene-Disease Validity and Clinical Actionability.

See New Dosage Map

ClinGen is seeking the genetics and broader medical community's valuable feedback on our work, tools, and resources, our 10 min survey is open to ClinGen contributors, users of ClinGen resources and those new to our work - we look forward to your input.

Provide Feedback

Search By Gene Name

Symbol:

Or click on the following examples: ZEB2, PTEN, MAPT

Go

Search By Location (GRCh37, GRCh38)

GRCh37: GRCh38:

Location: chr8:119,911,864-122,847,

Examples: chr2:44,000,000-45,500,000, 2p21-2p16.2

Go

Links

- ClinGen Home Page
- Help with this site
- FAQ
- Contact Us
- Curation of Recurrent CNVs
- Curation of the ACMG 59 Genes
- FTP

Submitted location: chr8:119,911,864-122,847,565

"N/A" indicates that this gene has not yet been evaluated

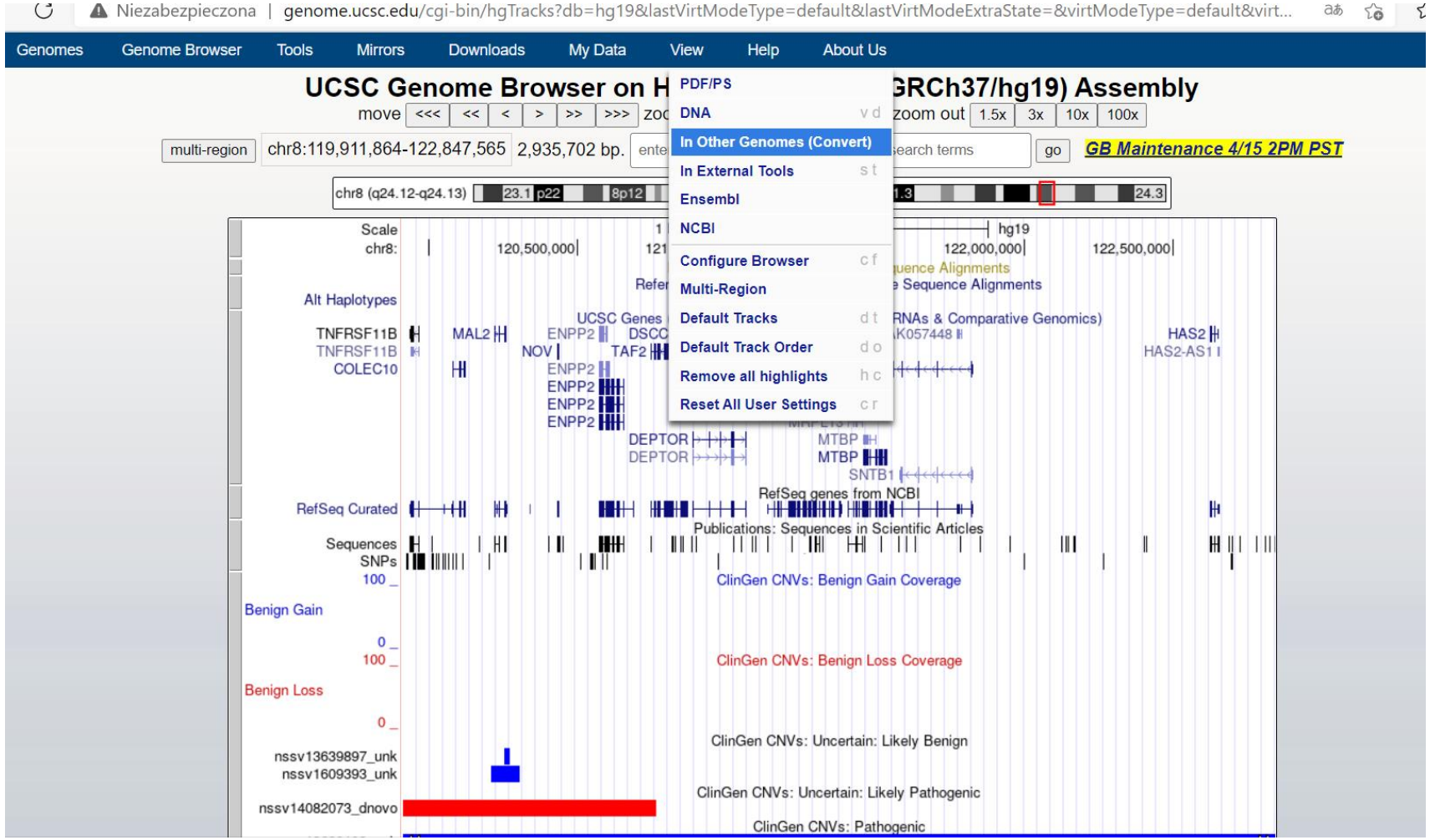
Gene Issues (29) | Region Issues (0)

Location search results

Items 1 - 20 of 29 < Prev Page 1 of 2 Next >

Gene Symbol	Haploinsufficiency score	Triplosensitivity score	Curation Status	Region Location (GRCh37)	ExAC pLI	OMIM	Relationship to Submitted Location	ISCA ID
TNFRSF11B	30: Gene associated with autosomal recessive phenotype	Not yet evaluated	Complete	chr8:119,935,796-119,964,383	0.272	602643	Contained	ISCA-20688
TAF2	30: Gene associated with autosomal recessive phenotype	0	Complete	chr8:120,743,014-120,845,074	0.566	604912	Contained	ISCA-6904
COLEC10	N/A	N/A	Awaiting Review	chr8:119,964,406-120,120,694	0.099	607620	Contained	ISCA-13893
RNU6-12P	N/A	N/A	Awaiting Review	chr8:119,988,752-119,988,857			Contained	ISCA-40431
LOC100286746	N/A	N/A	Awaiting Review	chr8:119,994,249-119,995,180			Contained	ISCA-26370
LOC101927513	N/A	N/A	Awaiting Review	chr8:120,075,181-120,081,021			Contained	ISCA-41064
MAL2	N/A	N/A	Awaiting Review	chr8:120,220,610-120,257,913	0.459	609684	Contained	ISCA-12713
MIR548AZ	N/A	N/A	Awaiting Review	chr8:120,337,411-120,337,505			Contained	ISCA-38163
NOV	N/A	N/A	Awaiting Review	chr8:120,428,552-120,436,678	0.114	164958	Contained	ISCA-23866

Konwertowanie pomiędzy bazami



Niezabezpieczona | genome.ucsc.edu/cgi-bin/hgConvert?hgsid=1087230757_xxjEkIhopkiUXORWSkZRFMaAtZkR&db=hg19

Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | Projects | Help | About Us

Convert chr8:119911864-122847565 to New Assembly

Old Genome:	Old Assembly:	New Genome:	New Assembly:	
Human	Feb. 2009 (GRCh37/hg19)	Human	Dec. 2013 (GRCh38/hg38)	Submit

Dec. 2013 (GRCh38/hg38)
Mar. 2006 (NCBI36/hg18)
May 2004 (NCBI35/hg17)

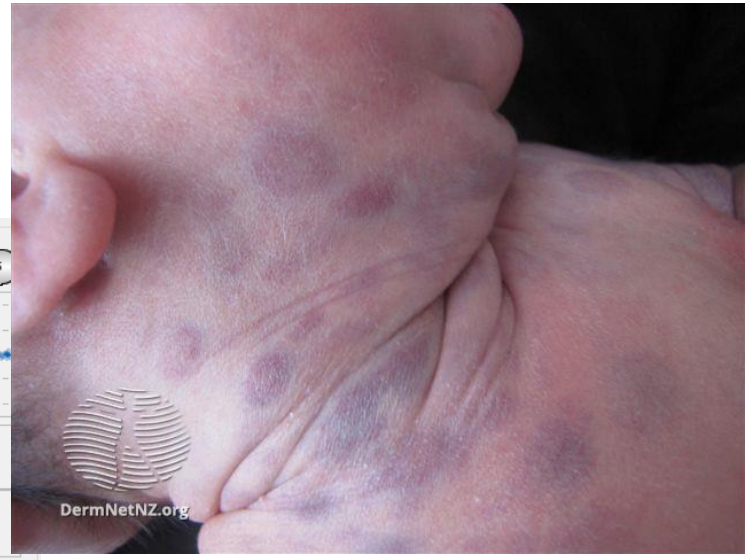
Niezabezpieczona | genome.ucsc.edu/cgi-bin/hgConvert?hgsid=1087230757_xxjEkIhopkiUXORWSkZRFMaAtZkR&hglft_toOrg=Human&hglft_toDb=h...

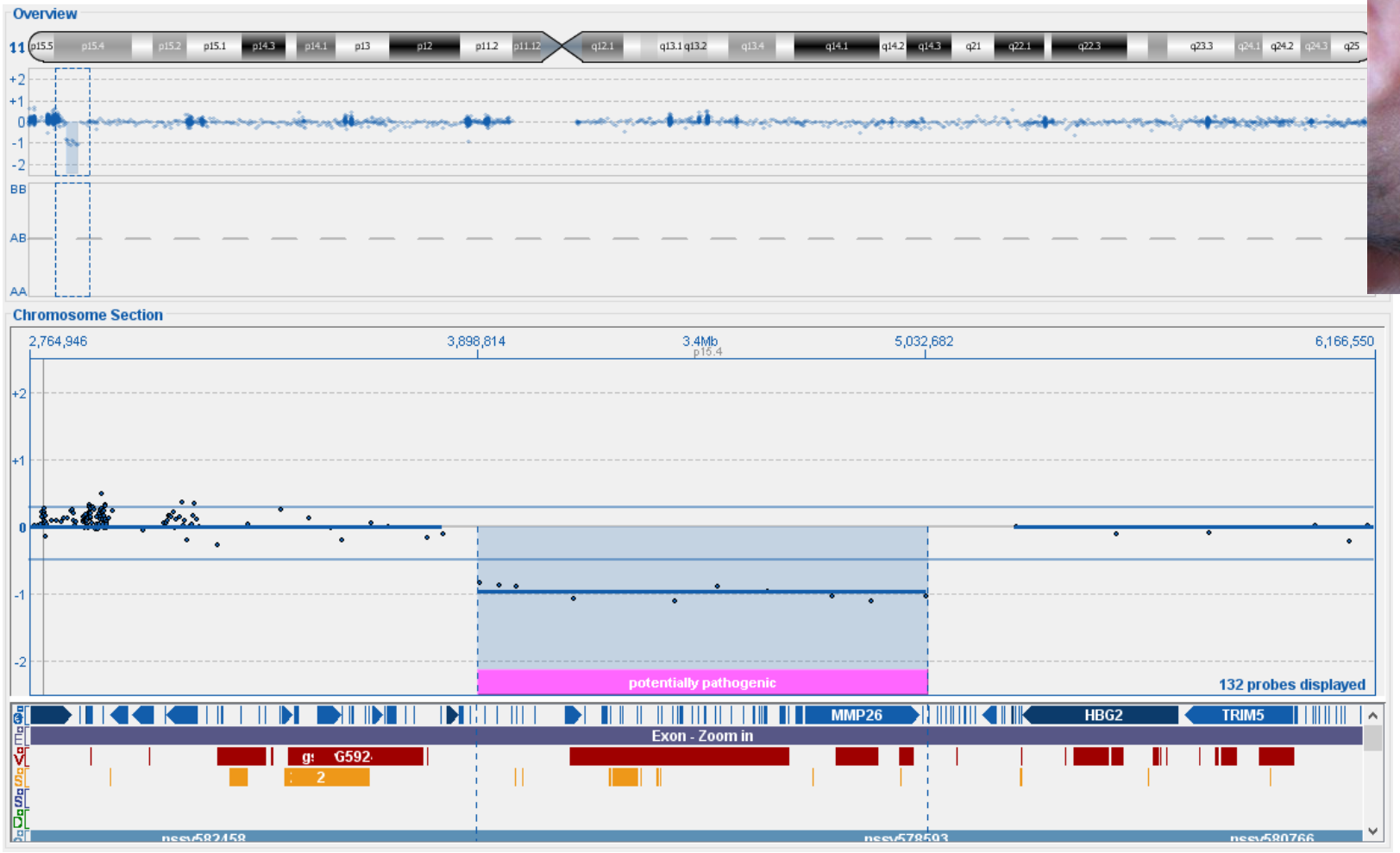
Genomes | Genome Browser | Tools | Mirrors | Downloads | My Data | Projects | Help | About Us

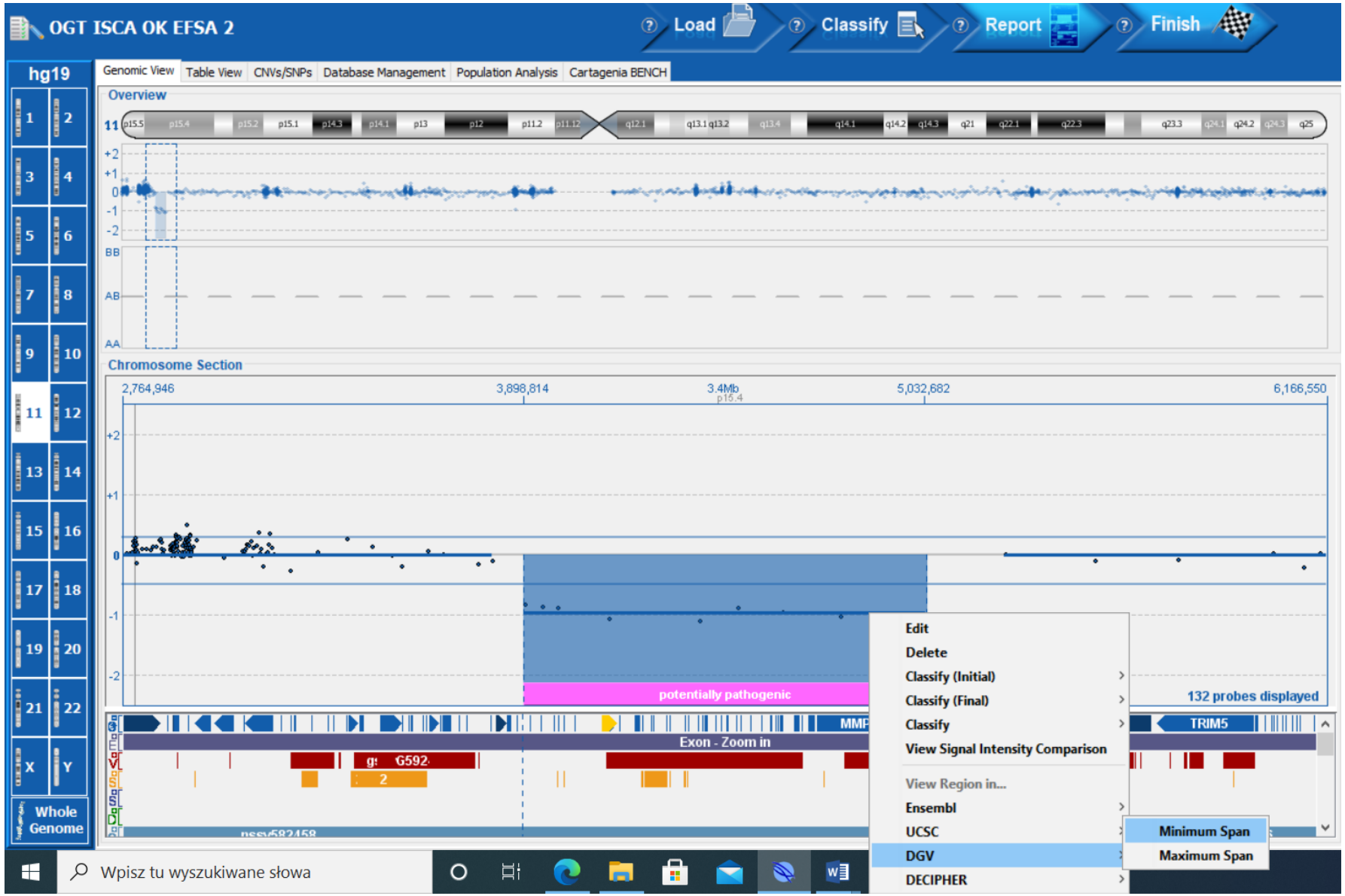
Human Feb. 2009 (GRCh37/hg19) chr8:119911864-122847565 to Human Dec. 2013 (GRCh38/hg38)

chr8:118899625-121835325 (100.0% of bases, 100.0% of span)

2-dniow noworodek, chłopiec, spodziectwo, blueberry muffin baby syndrome.
arr[hg19] 11p15.4(3,898,814-5,032,682)x1 ~1,3 Mpz






OGT ISCA OK EFSA 2
Load
Classify
Report
Finish
hg19
Genomic View
Table View
CNVs/SNPs
Database Management
Population Analysis
Cartagenia BENCH
Overview
Chromosome Section
2,764,946
3,898,814
3.4Mb
5,032,682
6,166,550
potentially pathogenic
132 probes displayed
Exon - Zoom in
TRIM5
Edit
Delete
Classify (Initial)
Classify (Final)
Classify
View Signal Intensity Comparison
View Region in...
Ensembl
UCSC
DGV
DECIPHER
Minimum Span
Maximum Span
Whole Genome
Wpisz tu wyszukiwane słowa

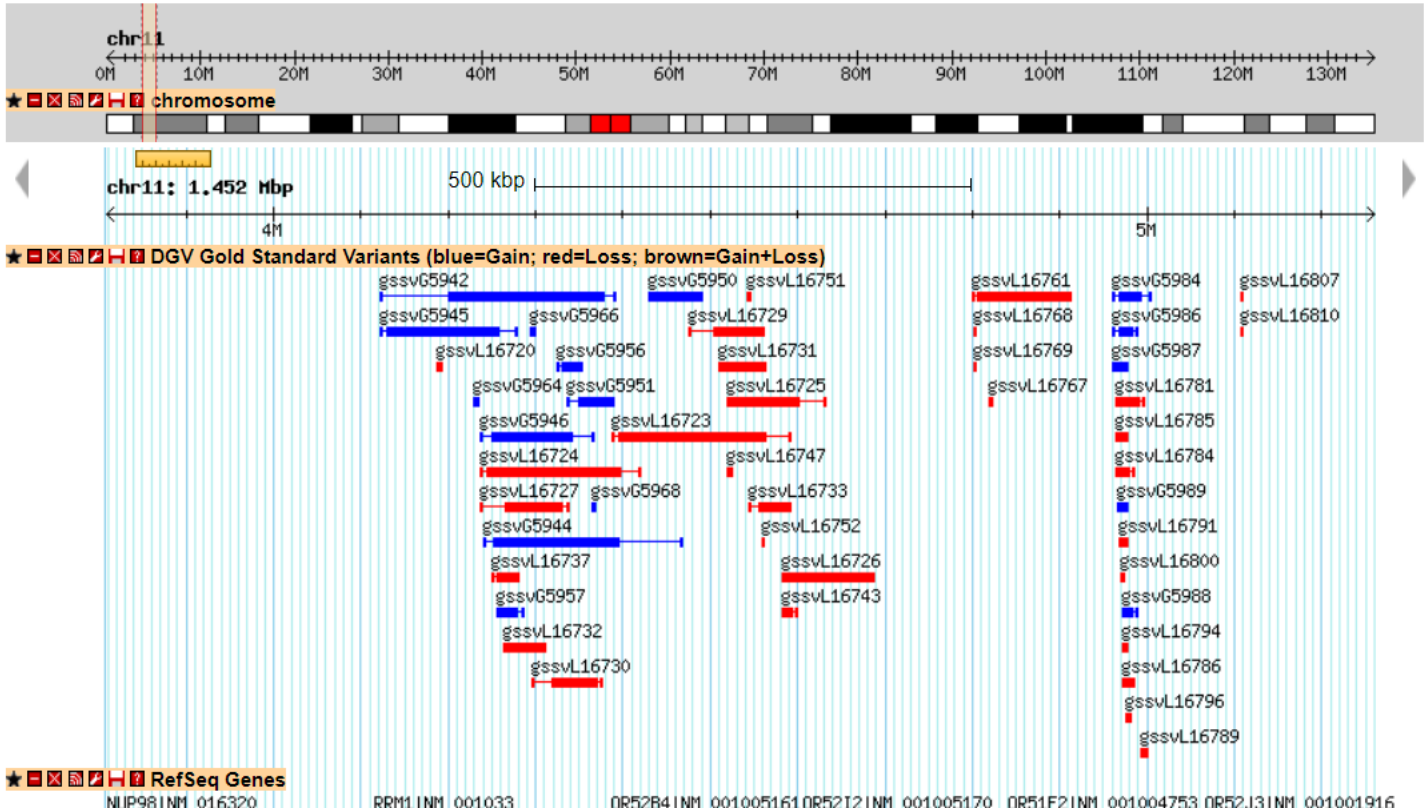
lub fragment:
:3,808,940..5,260,467 Szukaj
ady: chr7:71890181..72690180, CFTR, AC108171.3, nsv529033.
danych
mic Variants in Human Genome (Build GRCh37: Feb. 2009, hg19)

Przewijanie/powiększenie: Widoczno➢ć 1.452 Mbp Skieruj oś przeciwnie

variants
= + -
Reset

ąd

góły



Our URL has changed to https://www.deciphergenomics.org.
Links to https://decipher.sanger.ac.uk/ will continue to be redirected.

DECIPHER GRCh38 About Browse DDD (UK) Search DECIPHER Help Join Log in

Search results for "11:3877584-5011452" (Refine Search)

Patient variants 62 | CNV syndrome variants 0 | DDD research variants 5 | Genes 59

Genes: 1 to 10 of 33 (out of 59 total)

Show: OMIM Morbid DDG2P ClinGen Protein coding Filter...

Name / Description	Location	pLI	LOEUF	sHet	%HI	OMIM / Morbid	DDG2P	ClinGen	Links
RRM1 ribonucleotide reductase catalytic subunit M1	11 4094707 4138932	1.00	0.10	0.351	2.49	OMIM	-	-	View
STIM1 stromal interaction molecule 1	11 3854527 4093210	0.06	0.64	0.054	5.93	OMIM Morbid (3)	-	-	View
OR51E1 olfactory receptor family 51 subfamily E member 1	11 4643420 4655488	0.00	1.68	0.008	55.81	OMIM	-	-	View
OR51L1 olfactory receptor family 51 subfamily L member 1	11 4994851 5005536	-	-	0.015	63.18	-	-	-	View
OR51E2 olfactory receptor family 51	11 4680171 4697854	0.00	1.68	0.015	64.41	OMIM	-	-	View

Gain/Loss Loss ×

Size 1-10 Mb ×

- [ ] 0.01-0.1 Mb
- [ ] 0.1-1 Mb
- [x] 1-10 Mb
- [ ] 10-100 Mb
- [ ] Over 100 Mb

Gain/Loss

Loss: 100%

e

Maternally inherited: 33%

Imbalance arising from a balanced parental rearrangement: 33%

Inherited from parent with similar phenotype to child: 33%

Size

1-10 Mb: 100%

Phenotypes present in multiple matching patients

No phenotypes are present in multiple matching patients for the current filters

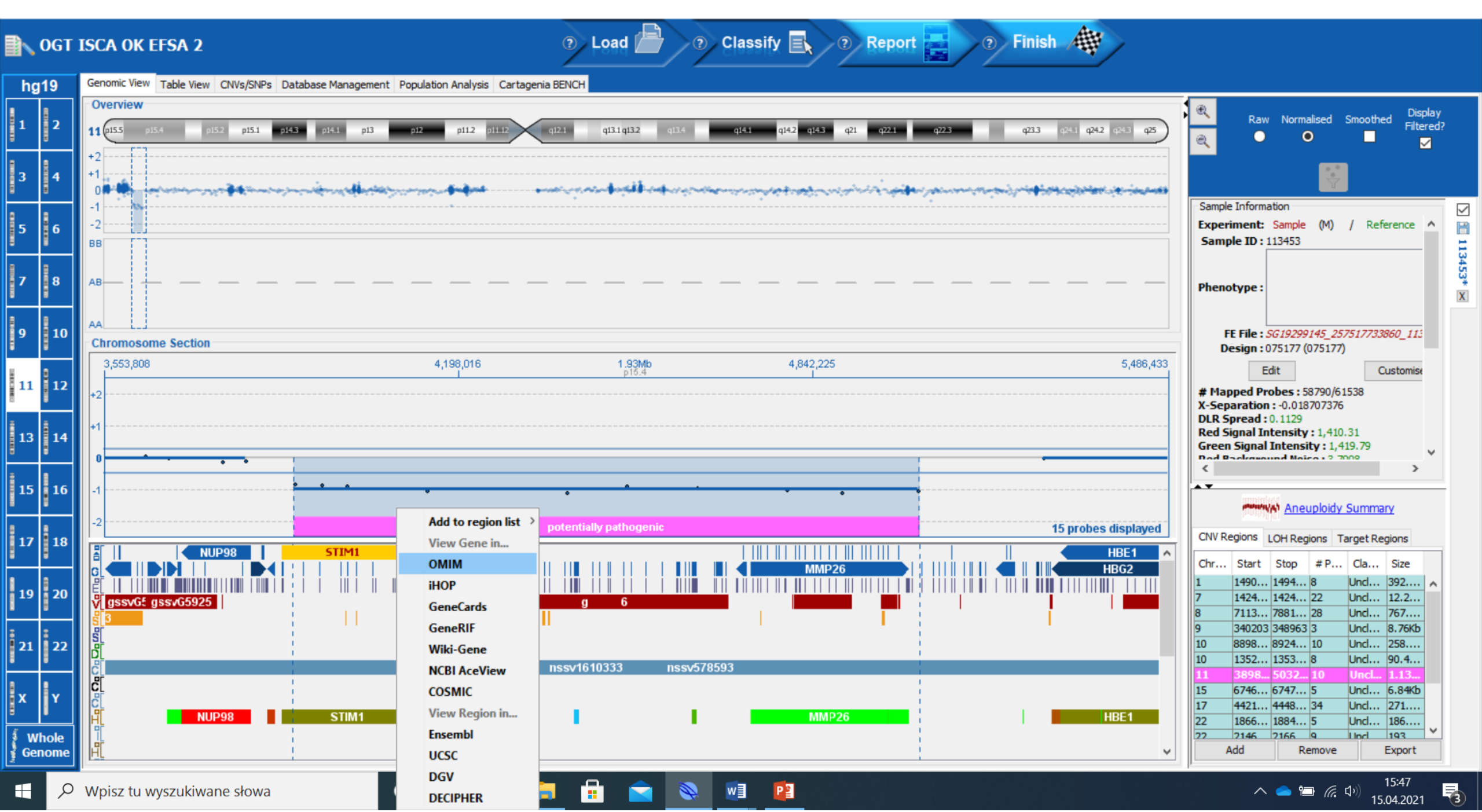
This patient's phenotypes present in matching patients

1 Abnormal erythrocyte morphology

This patient's phenotypes absent in matching patients

- Capillary hemangioma

Patient	Sex	Size	Overlap	Genes in common	Functional similarity	Type	Pathogenicity / Contribution	Inheritance / Genotype	Shared Phenotypes
4441	46XY	1.52 Mb	856.41 kb	64	0.58	Deletion		Inherited from parent with similar phenotype to child Heterozygous	1 of 1: **Abnormal erythrocyte morphology**
389893	46XX	2.06 Mb	197.72 kb	16	0.15	Deletion	Pathogenic Full	Imbalance arising from a balanced parental rearrangement Heterozygous	0 of 5: Global developmental delay; Hypotonia; Neonatal hypotonia; Generalized hypotonia; Fused labia minora
284239	46XY	2.36 Mb	34.63 kb	5	0.05	Deletion	Uncertain Partial	Maternally inherited Heterozygous	0 of 9: Short phalanx of finger; Webbed neck; Intellectual disability, severe; Low-set ears;


OGT ISCA OK EFSA 2
Load
Classify
Report
Finish
hg19
Genomic View
Table View
CNVs/SNPs
Database Management
Population Analysis
Cartagenia BENCH
Overview
Chromosome Section
3,553,808
4,198,016
1.93Mb
4,842,225
5,486,433
potentially pathogenic
15 probes displayed
Add to region list
View Gene in...
OMIM
iHOP
GeneCards
GeneRIF
Wiki-Gene
NCBI AceView
COSMIC
View Region in...
Ensembl
UCSC
DGV
DECIPHER
NUP98
STIM1
MMP26
HBE1
HBG2
nssv1610333
nssv578593
Raw
Normalised
Smoothed
Display Filtered?
Sample Information
Experiment: Sample (M) / Reference
Sample ID : 113453
Phenotype :
FE File : SG19299145_257517733860_113
Design : 075177 (075177)
Edit
Customise
Mapped Probes : 58790/61538
X-Separation : -0.018707376
DLR Spread : 0.1129
Red Signal Intensity : 1,410.31
Green Signal Intensity : 1,419.79
Aneuploidy Summary
CNV Regions
LOH Regions
Target Regions
Chr... Start Stop # P... Cla... Size
1 1490... 1494... 8 Uncl... 392....
7 1424... 1424... 22 Uncl... 12.2...
8 7113... 7881... 28 Uncl... 767....
9 340203 348963 3 Uncl... 8.76Kb
10 8898... 8924... 10 Uncl... 258....
10 1352... 1353... 8 Uncl... 90.4...
11 3898... 5032... 10 Uncl... 1.13...
15 6746... 6747... 5 Uncl... 6.84Kb
17 4421... 4448... 34 Uncl... 271....
22 1866... 1884... 5 Uncl... 186....
Add
Remove
Export
113453*
Whole Genome
Wpisz tu wyszukiwane słowa
15:47
15.04.2021

* 605921

STROMAL INTERACTION MOLECULE 1; STIM1

HGNC Approved Gene Symbol: STIM1

Cytogenetic location: 11p15.4 *Genomic coordinates (GRCh38): 11:3,854,603-4,093,209* (from NCBI)

Gene-Phenotype Relationships

Location	Phenotype Clinical Synopses	Phenotype MIM number	Inheritance	Phenotype mapping key
11p15.4	Immunodeficiency 10	612783	AR	3
	Myopathy, tubular aggregate, 1	160565	AD	3
	Stormorken syndrome	185070	AD	3

PheneGene Graphics

TEXT

Description

STIM1 is a calcium sensor that conveys the calcium load of the endoplasmic reticulum (ER) to store-operated channels (SOCs) at the plasma membrane (Yuan et al., 2007).

Cloning and Expression

External Links
Genome
DNA
Protein
Gene Info
Clinical Resources
Variation
1000 Genome
ClinVar
gnomAD
GWAS Catalog
GWAS Central
HGMD
HGVS
NHLBI EVS
PharmGKB
Animal Models
Cellular Pathways

WYNIK:

arr[GRCh37] 11p15.4(3898814_5032682)x1

INTERPRETACJA:

Stwierdzono niezrównoważenie genomu w postaci delecji krótkiego ramienia chromosomu 11 pary w regionie 11p15.4 o wielkości ~1,13 Mpz.

Delecja obejmuje 31 genów kodujących białka, w tym geny: *TRIM21* (OMIM 109092)[1], *TRIM68* (OMIM 613184)[1], *OR51E1* (OMIM 611267)[1], *OR51E2* (OMIM 611268)[1], *MMP26* (OMIM 605470)[1] oraz eksony 2-13 wrażliwego na dawkę genu *STIM1* (OMIM 605921)[1] i wrażliwy na dawkę gen *RRM1* (OMIM 180410)[1]. Mutacje w genie *STIM1* opisywane są u pacjentów z dziedziczonymi w sposób autosomalny dominujący zespołem Stormorkena (trombocytopatia - brak śledziony – mioza) (STRMK, OMIM 185070)[1] i miopatią z tubularnymi agregatami (TAM1, OMIM 160565)[1] oraz dziedziczonym w sposób autosomalny recesywny złożonym niedoborem odporności z powodu deficytu STIM1 (IMD10, OMIM 612783)[1]. U pacjentów z zespołem Stormorkena opisywane są zmiany skórne (plamica, rybia łuska)[4].

W literaturze opisano pacjenta z epsilon-gamma-delta-beta talasemią, u którego stwierdzono patogenną delecję w regionie 11p15.4 o wielkości ~2,2 Mpz (chr11:4073950_6274517), obejmującą klaster genów β-globiny: *HBE1, HBG1, HBG2, HBD* i *HBB* oraz eksony 4-12 genu *STIM1*[2]. Epsilon-gamma-delta-beta talasemię opisano również u pacjenta ze zmianami o charakterze *blueberry muffin*, u którego stwierdzono delecję w regionie 11p15.4 o wielkości ~59 kpz, obejmującą 3 geny z klastra genów β-globiny[3].

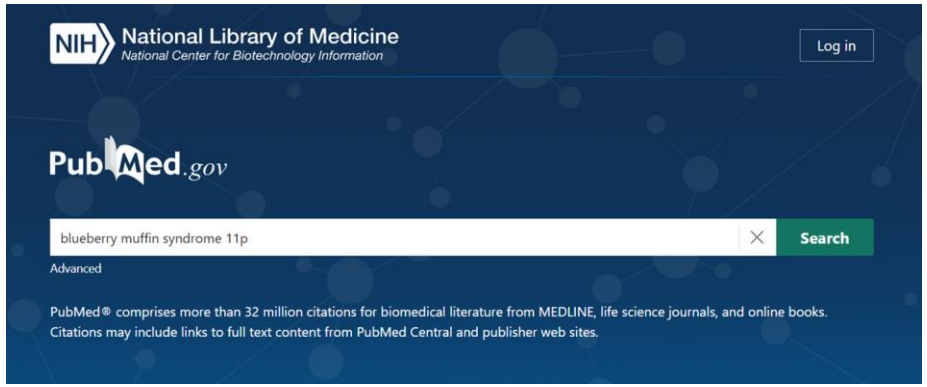
Aberracja należy do zmian o charakterze potencjalnie patogennym.

ZALECENIA:

Wskazana konsultacja w Poradni Genetycznej.
W celu pełnej interpretacji wyniku badania, ustalenia pochodzenia stwierdzonej aberracji oraz określenia ryzyka genetycznego w rodzinie należy wykonać badania metodą FISH u pacjenta i jego rodziców. W tym celu należy pobrać od nich krew na heparynę.

ŹRÓDŁA INFORMACJI:

1. https://omim.org
2. Repnikova E, Roberts J, Mc Dermott S, Farooqi MS, Iqbal NT, Silvey M, Nolen J, Taboada E, Li W. Clinical and mole characterization of novel deletions causing epsilon gamma delta beta thalassemia: Report of two cases. Pathol Res Pract Oct;215(10):152578.



Case Reports › Pediatr Dev Pathol. Nov-Dec 2019;22(6):599-600.
doi: 10.1177/1093526619850663. Epub 2019 May 14.

Blueberry Muffin Skin Lesions in an Infant With Epsilon Gamma Delta Beta Thalassemia

Neha Puar [1], Brandon Newell [2], Lei Shao [3]

Affiliations + expand

PMID: 31088202 DOI: 10.1177/1093526619850663

Fundusze Europejskie Wiedza Edukacja Rozwój
Rzeczpospolita Polska
Instytut Matki i Dziecka
Unia Europejska Europejski Fundusz Społeczny
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orphanet

Pomoc Drukuj Skontakt

Portal dla **rzadkich chorób** i **leków sierocych**

COVID-19 & Choroby rzadkie orphanet

Znajdź zalecenia i usługi oferowane przez ekspertów dotyczące COVID-19 i chorób rzadkich, w tym te, które oferują Europejskie Sieci Referencyjne (European Reference Networks), są one dostępne w wielu różnych językach.

Choroby rzadkie

- Wyszukaj
- Oznaki i objawy kliniczne
- Klasyfikacje
- Geny
- Niepełnosprawność
- Encyklopedia dla pacjentów
- Encyklopedia dla specjalistów
- Przewodniki postępowania
- Źródła / procedury

Strona główna > Choroby rzadkie > **Wyszukaj**

Wyszukaj **chorobę rzadką**

TANGO2 *

Szukaj

(*) pole obowiązkowe

- ○ Wyszukaj według nazwy choroby
- ○ Kod ORPHA
- ○ OMIM
- ○ ICD-10
- ◉ Nazwa lub symbol genów

Inne opcje wyszukiwania

Choroby rzadkie

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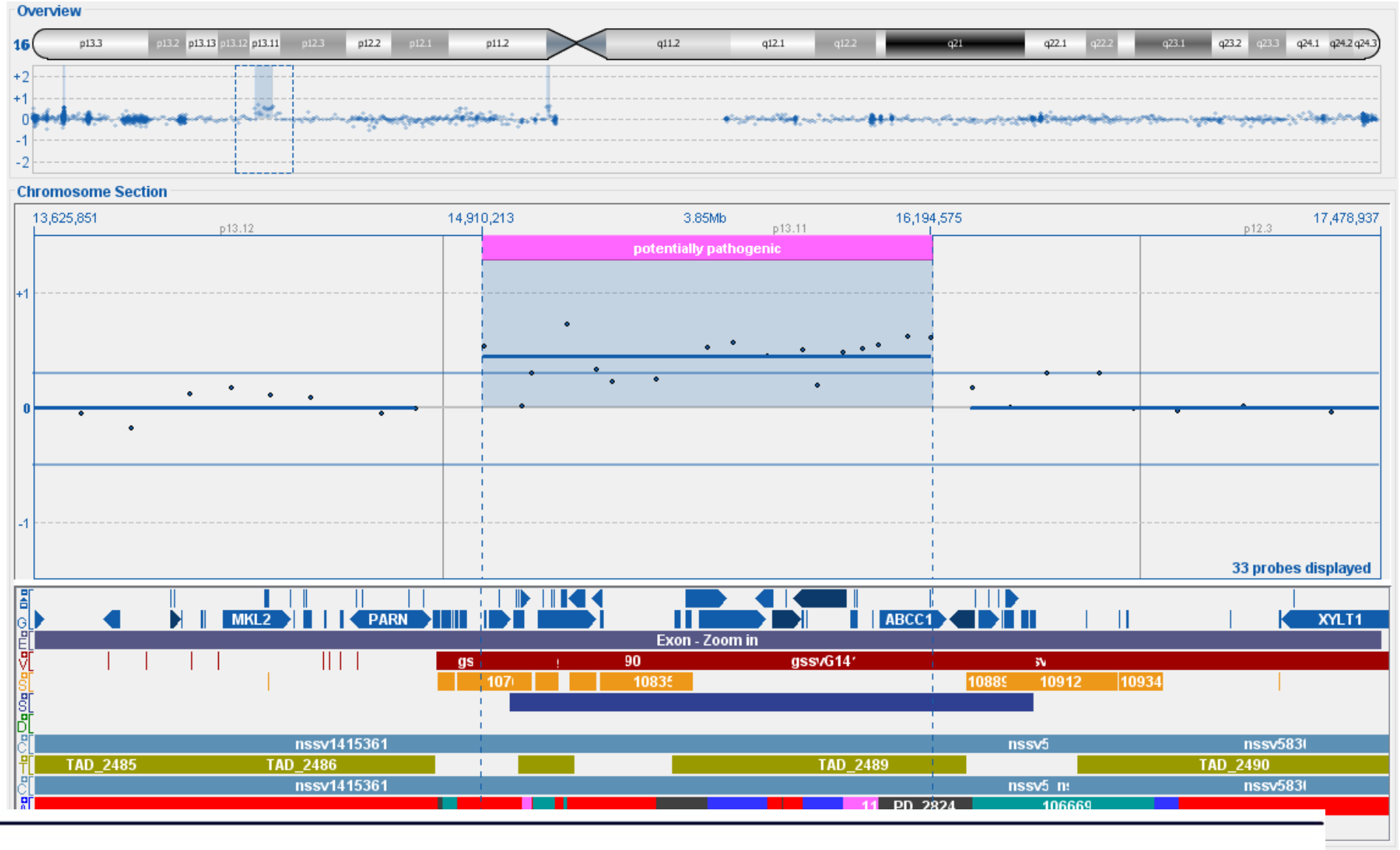
Inne opcje wyszukiwania

1 Wyniki

ORPHA:480864 () Zespół nawracającego przełomu metabolicznego o charakterze encefalomiopatii, rabdomiolizy, arytmii serca i niepełnosprawności intelektualnej

Więcej informacji

3 – letni chłopiec. Wskazania: zespół wad wrodzonych, hipotrofia, niska odporność
arr[hg19] 16p13.11(14,910,213-16,194,575)x3 1.6Mb – **region o niepełnej penetracji**



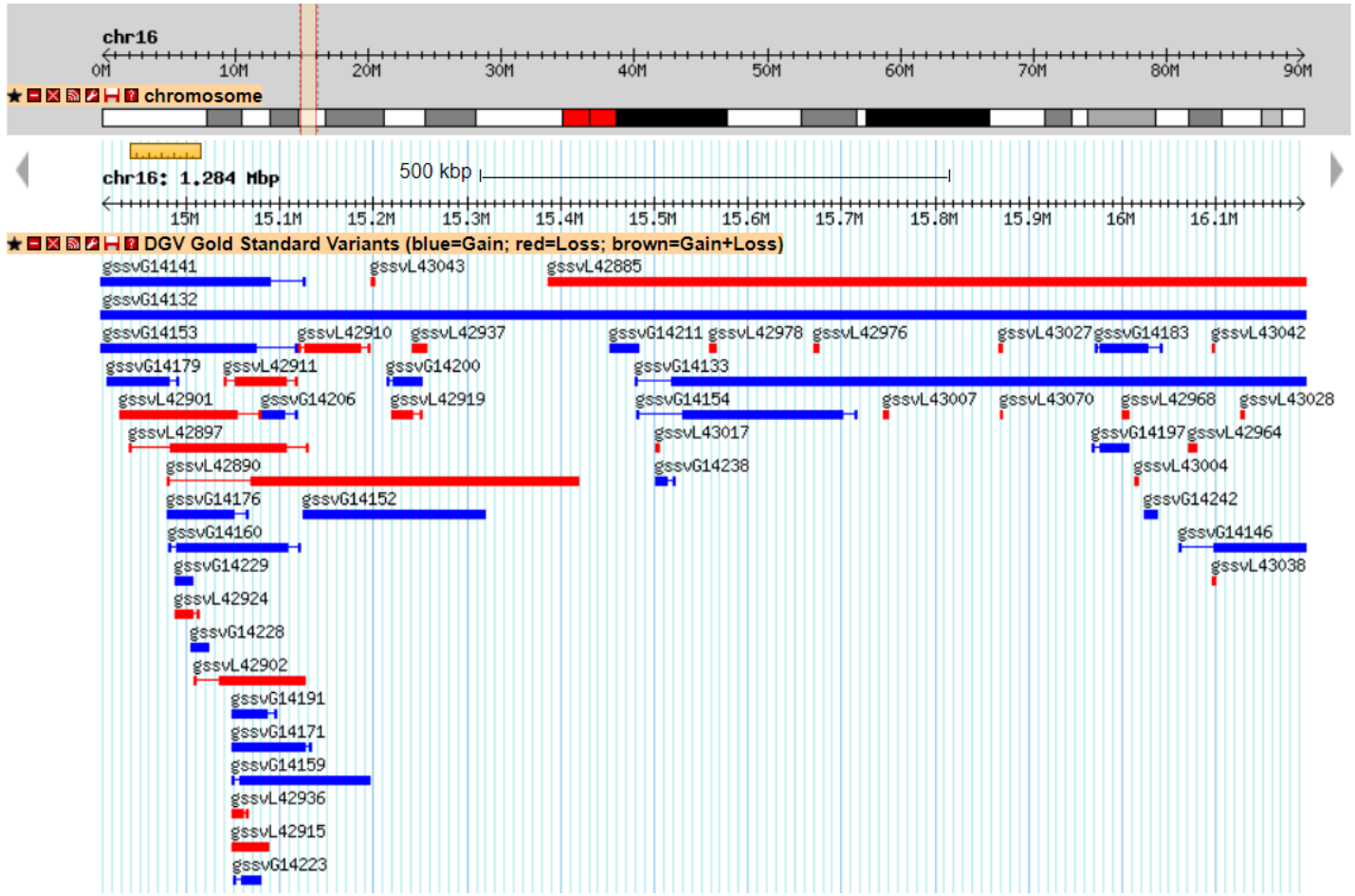
Filter variants

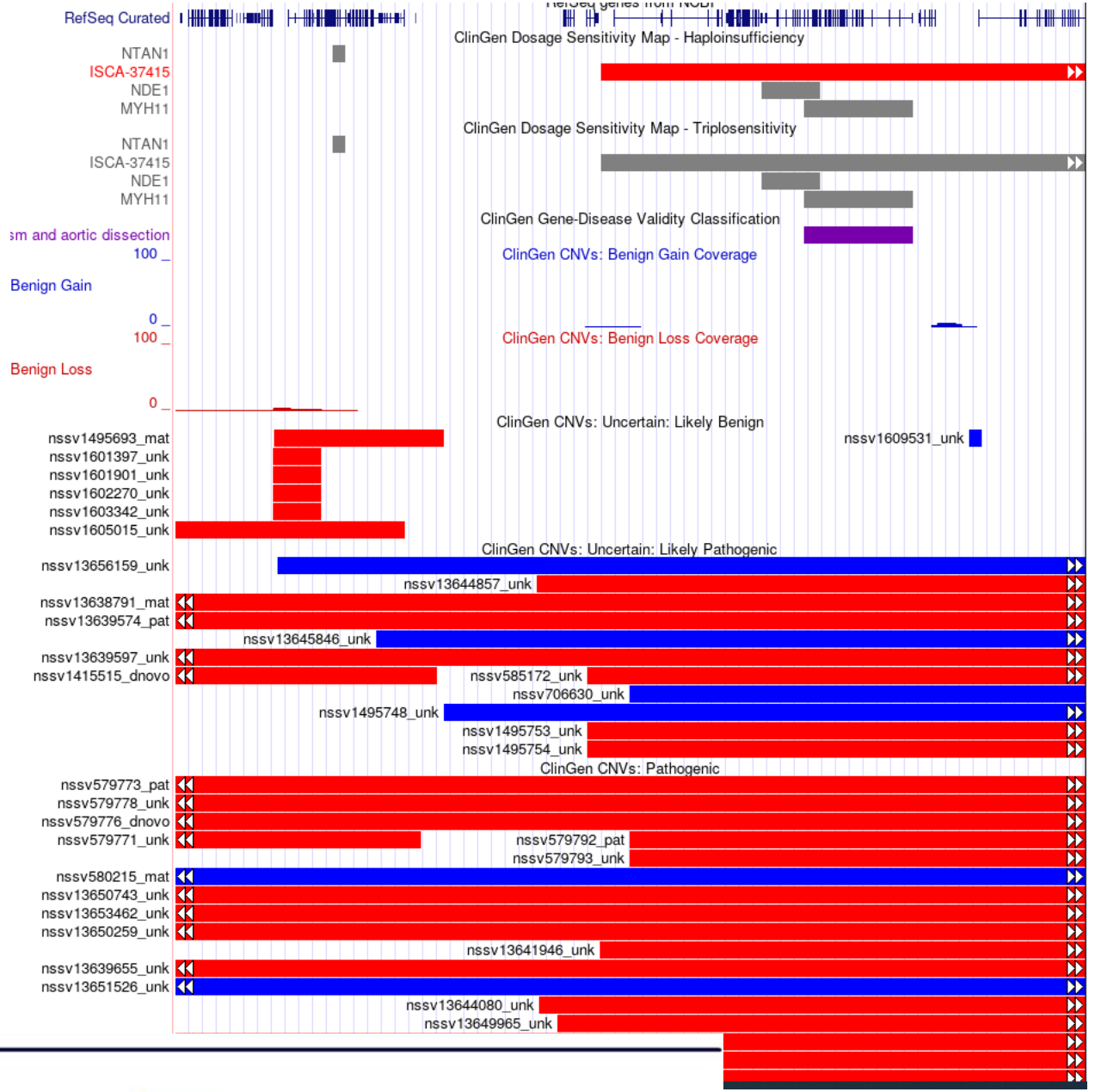
study = + -

Filter Reset

Przegląd

Szczegóły




RefSeq Curated
ClinGen Dosage Sensitivity Map - Haploinsufficiency
NTAN1
ISCA-37415
NDE1
MYH11
ClinGen Dosage Sensitivity Map - Triplosensitivity
NTAN1
ISCA-37415
NDE1
MYH11
ClinGen Gene-Disease Validity Classification
sm and aortic dissection
ClinGen CNVs: Benign Gain Coverage
100
Benign Gain
0
ClinGen CNVs: Benign Loss Coverage
100
Benign Loss
0
ClinGen CNVs: Uncertain: Likely Benign
nssv1495693_mat
nssv1609531_unk
nssv1601397_unk
nssv1601901_unk
nssv1602270_unk
nssv1603342_unk
nssv1605015_unk
ClinGen CNVs: Uncertain: Likely Pathogenic
nssv13656159_unk
nssv13644857_unk
nssv13638791_mat
nssv13639574_pat
nssv13645846_unk
nssv13639597_unk
nssv1415515_dnovo
nssv585172_unk
nssv706630_unk
nssv1495748_unk
nssv1495753_unk
nssv1495754_unk
ClinGen CNVs: Pathogenic
nssv579773_pat
nssv579778_unk
nssv579776_dnovo
nssv579771_unk
nssv579792_pat
nssv579793_unk
nssv580215_mat
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nssv13639655_unk
nssv13651526_unk
nssv13644080_unk
nssv13649965_unk

Dziękuję za uwagę

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