BioDiscovery Nexus Training Course

Nexus Copy Number Training

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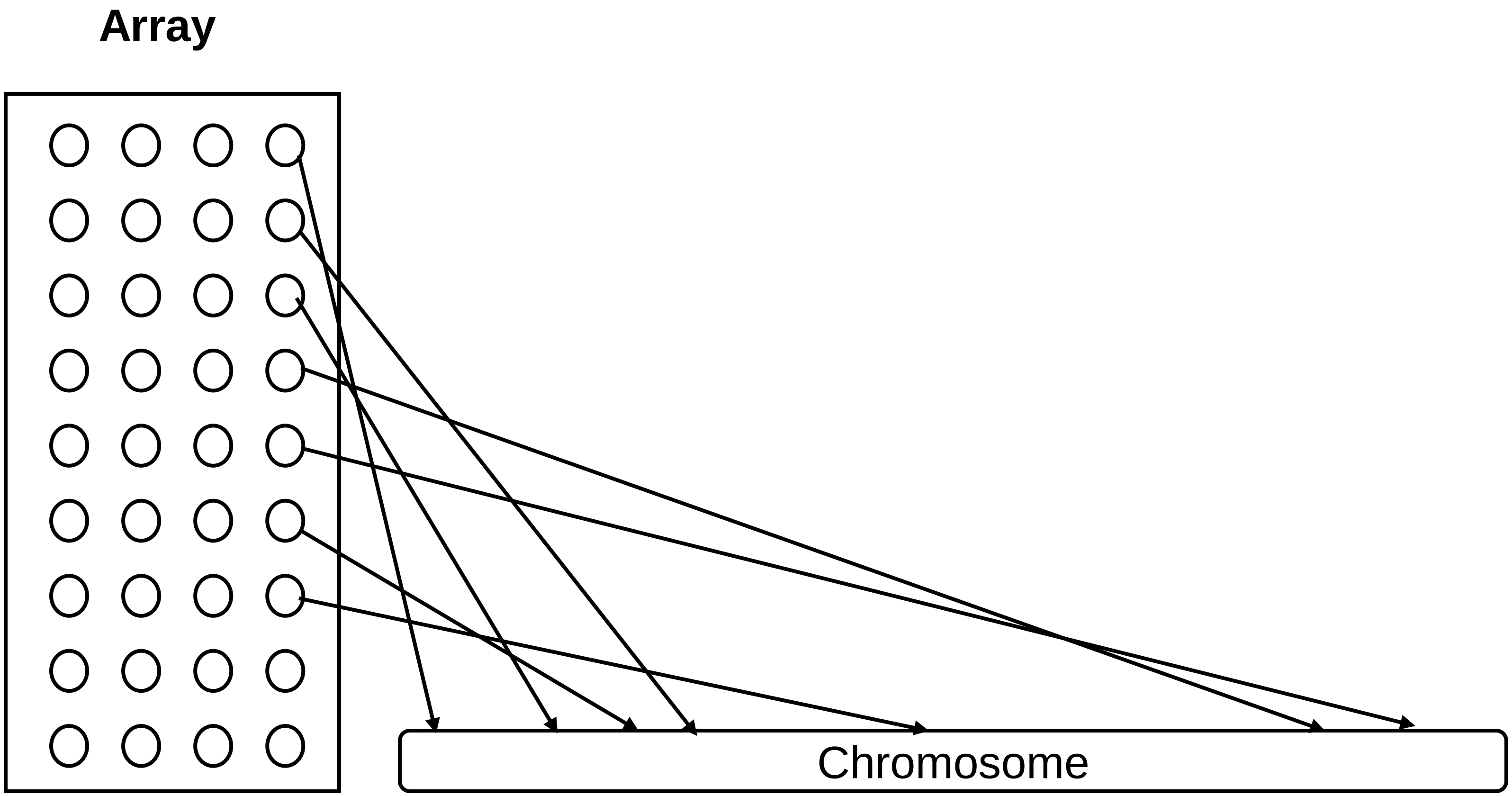
BioDiscovery, Inc.

Agenda

- Copy number analysis overview
- Downstream analysis
    - Visual inspection for changes of interest (chromothripsis) and ploidy
    - Identifying frequent and significant peaks (STAC, GISTIC)
    - Identifying significant co-occurring alterations
    - Comparing groups to find significant differences
- Nexus DB
    - Querying and downloading TCGA data
- Project set-up and processing
    - Create Project
    - Process samples
    - Add annotation

Copy number analysis overview

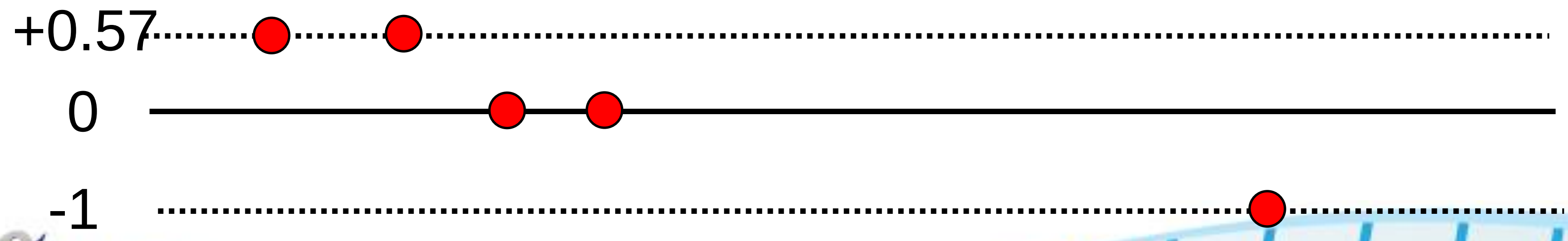
Mapping of probes to genome


Array
Chromosome

Plotting the Log Ratios

- Experiment results in a table (Two-color array like Agilent):

Probe Location	Expr.	Control	Ratio	Log Ratio
Chr1:10-20	150	100	3/2	+0.57
Chr1:50-60	300	200	3/2	+0.57
Chr1:70-90	500	500	2/2	0
Chr1:100-120	60	60	2/2	0
Chr1:250-300	500	1000	1/2	-1



SNP Array Data

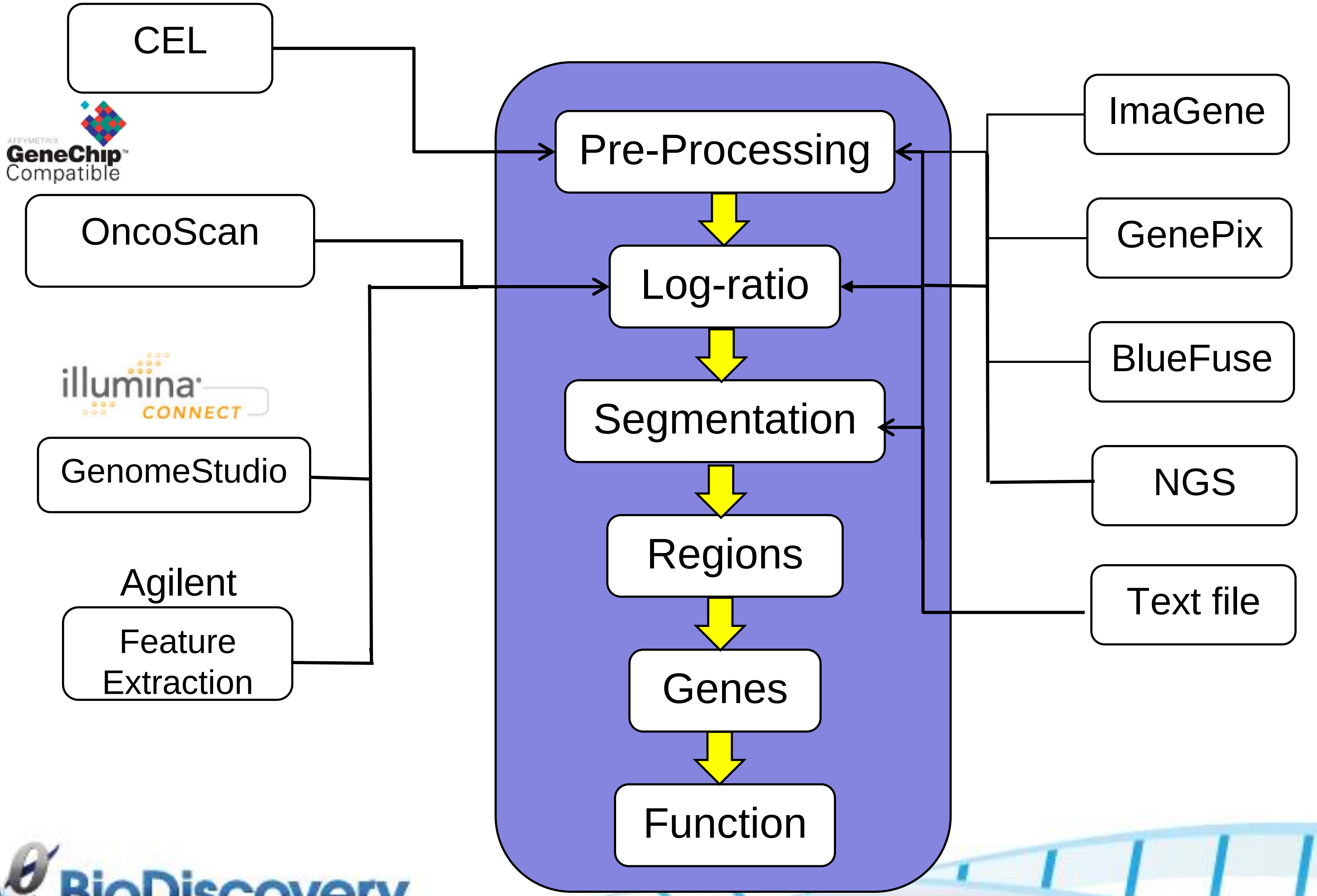
- SNP arrays use short oligos to interrogate a single SNP. However, the signal strength from the probe can be used for Copy Number estimation

- SNP Arrays are single color but a pool of arrays can be used to form a “reference” intensity value for a probe

- These platforms can also determine the zygosity of the probe as AA, AB, or BB

- Provided by Affymetrix and Illumina

Nexus Copy Number Workflow



CEL

GeneChip Compatible

OncoScan

illumina CONNECT

GenomeStudio

Agilent

Feature Extraction

Pre-Processing

Log-ratio

Segmentation

Regions

Genes

Function

ImaGene

GenePix

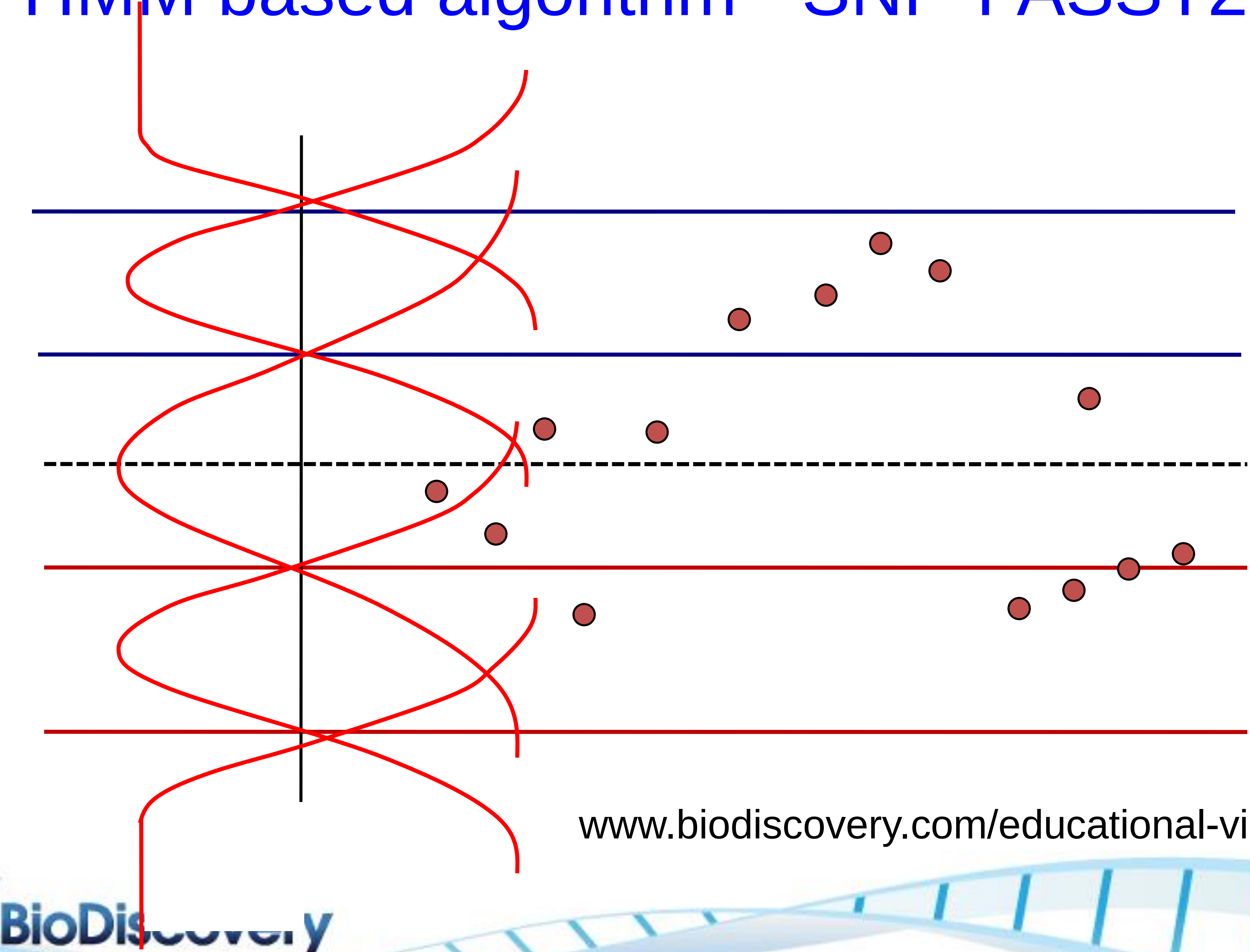
BlueFuse

NGS

Text file

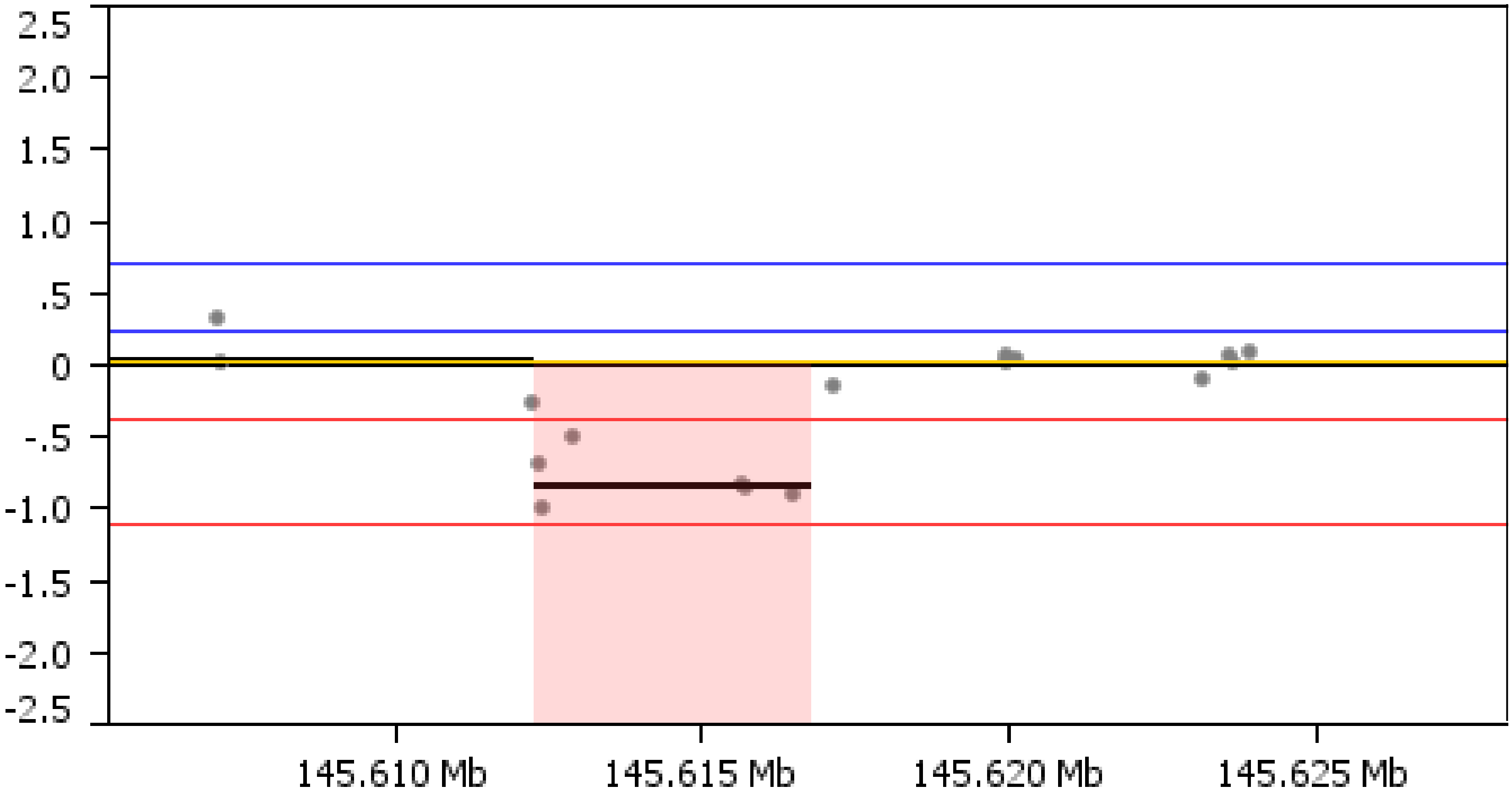
BioDiscovery

HMM based algorithm - SNP-FASST2

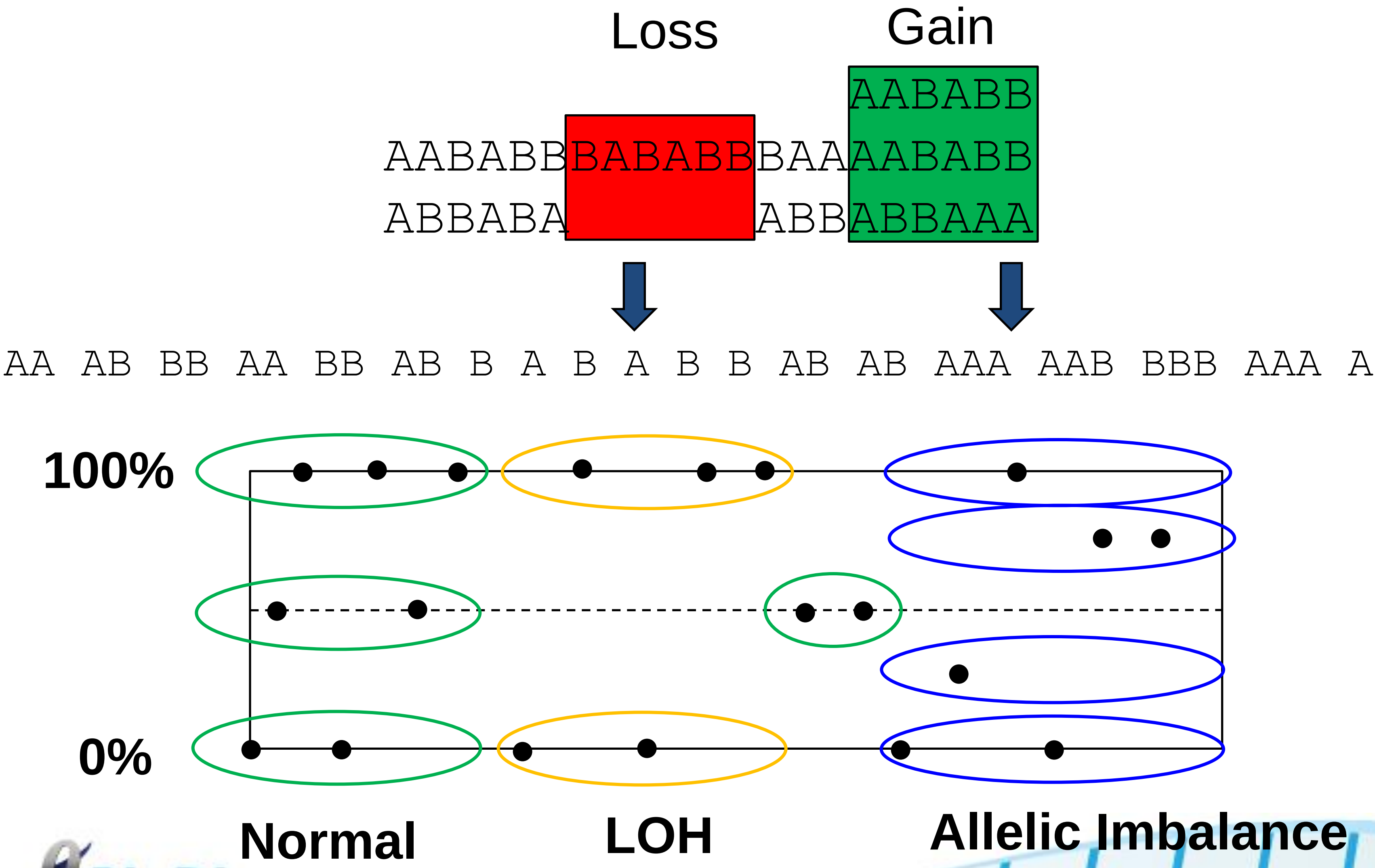


www.biodiscovery.com/educational-videos

Segmenting the Probes



B-Allele Freq. Bands



Loss
Gain

AABABB
AABABBBABABBBAAAABABB
ABBABA ABBABBAAA

AA AB BB AA BB AB B A B A B B AB AB AAA AAB BBB AAA ABB ABB

Normal
LOH
Allelic Imbalance

TCGA Premier and Nexus DB

Nexus DB – Web-based Repository of Nexus Projects

- Secure and redundant back-up and archive projects

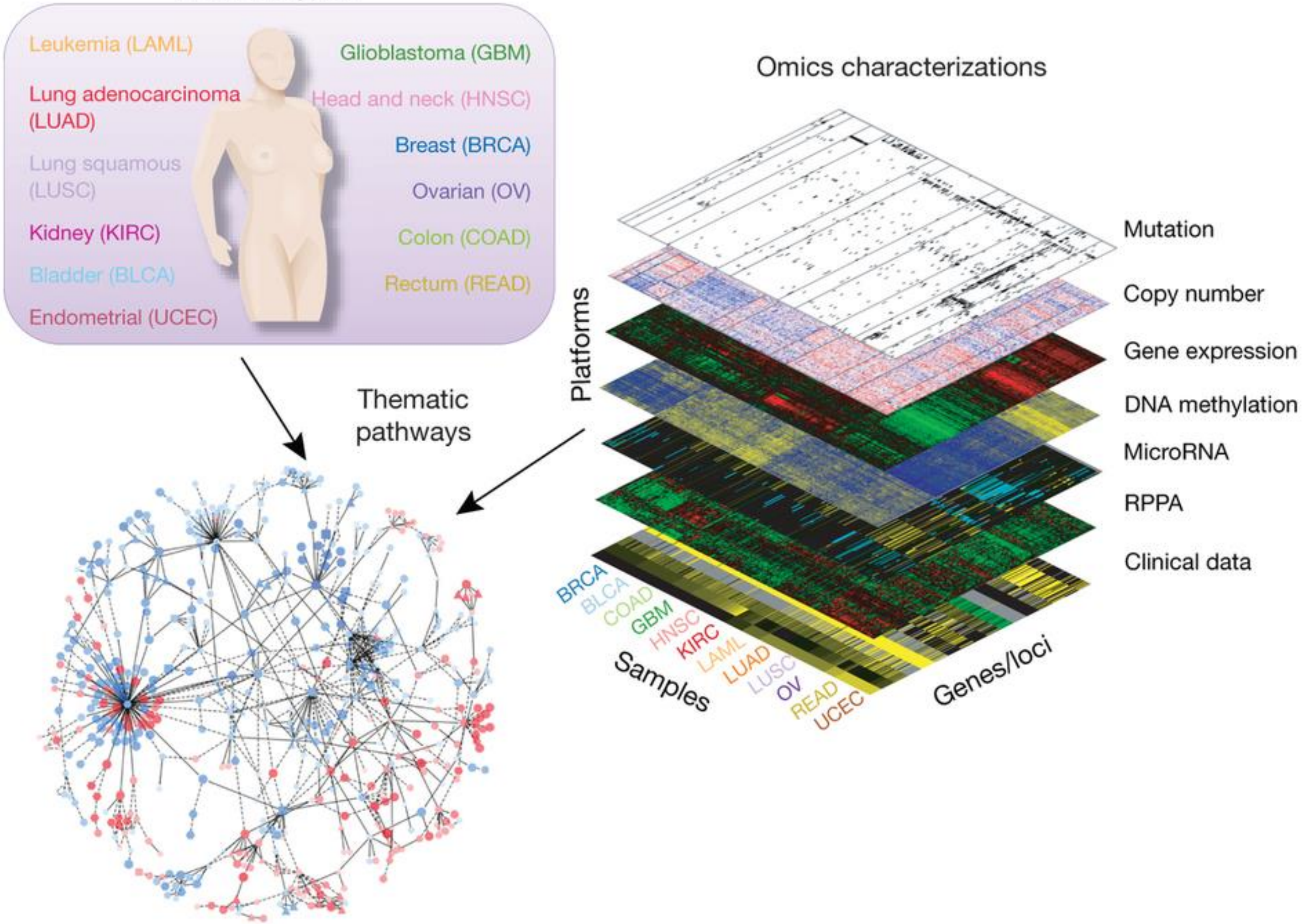
- Allow for queries across multiple project
  - Based on sample phenotype or genotype

- Download datasets that are ready to be analyzed immediately (e.g. TCGA, etc.)

- Share projects with collaborators

The Cancer Genome Atlas (TCGA)

26 Tumor Types (+6)

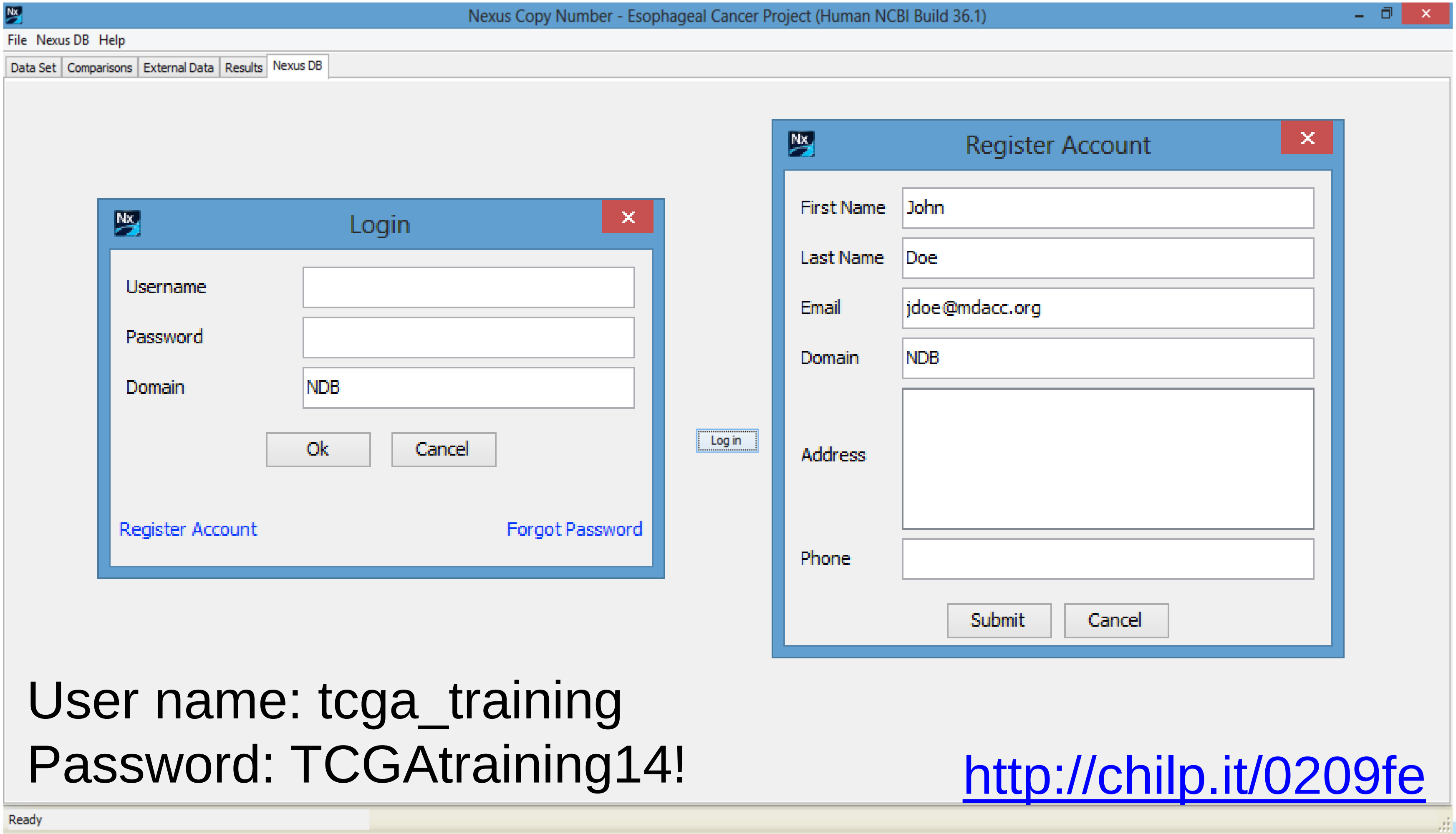

Leukemia (LAML)
Lung adenocarcinoma (LUAD)
Lung squamous (LUSC)
Kidney (KIRC)
Bladder (BLCA)
Endometrial (UCEC)
Glioblastoma (GBM)
Head and neck (HNSC)
Breast (BRCA)
Ovarian (OV)
Colon (COAD)
Rectum (READ)
Thematic pathways
Omics characterizations
Platforms
Mutation
Copy number
Gene expression
DNA methylation
MicroRNA
RPPA
Clinical data
BRCA
BLCA
COAD
GBM
HNSC
KIRC
LAML
LUAD
LUSC
OV
READ
UCEC
Samples
Genes/loci

TCGA Data in Nexus DB

- Nexus project has been created for each cancer type
  - Level 3 (data processed by TCGA pipeline)
  - Level 1 (reprocessed and curated for ploidy and QC)

- Nexus Copy Number users can:
  - Query this data to identify samples and frequency of events meeting query criteria
  - Download any of the samples to a local Nexus Copy Number project to integrate with other data or by themselves and perform aggregate analysis

Nexus DB Access



Nexus Copy Number - Esophageal Cancer Project (Human NCBI Build 36.1)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Results
Nexus DB
Login
Ok
Cancel
Register Account
Forgot Password
Log in
Register Account
Submit
Cancel
Ready

Login

Username:

Password:

Domain: NDB

Register Account

First Name: John

Last Name: Doe

Email: jdoe@mdacc.org

Domain: NDB

Address:

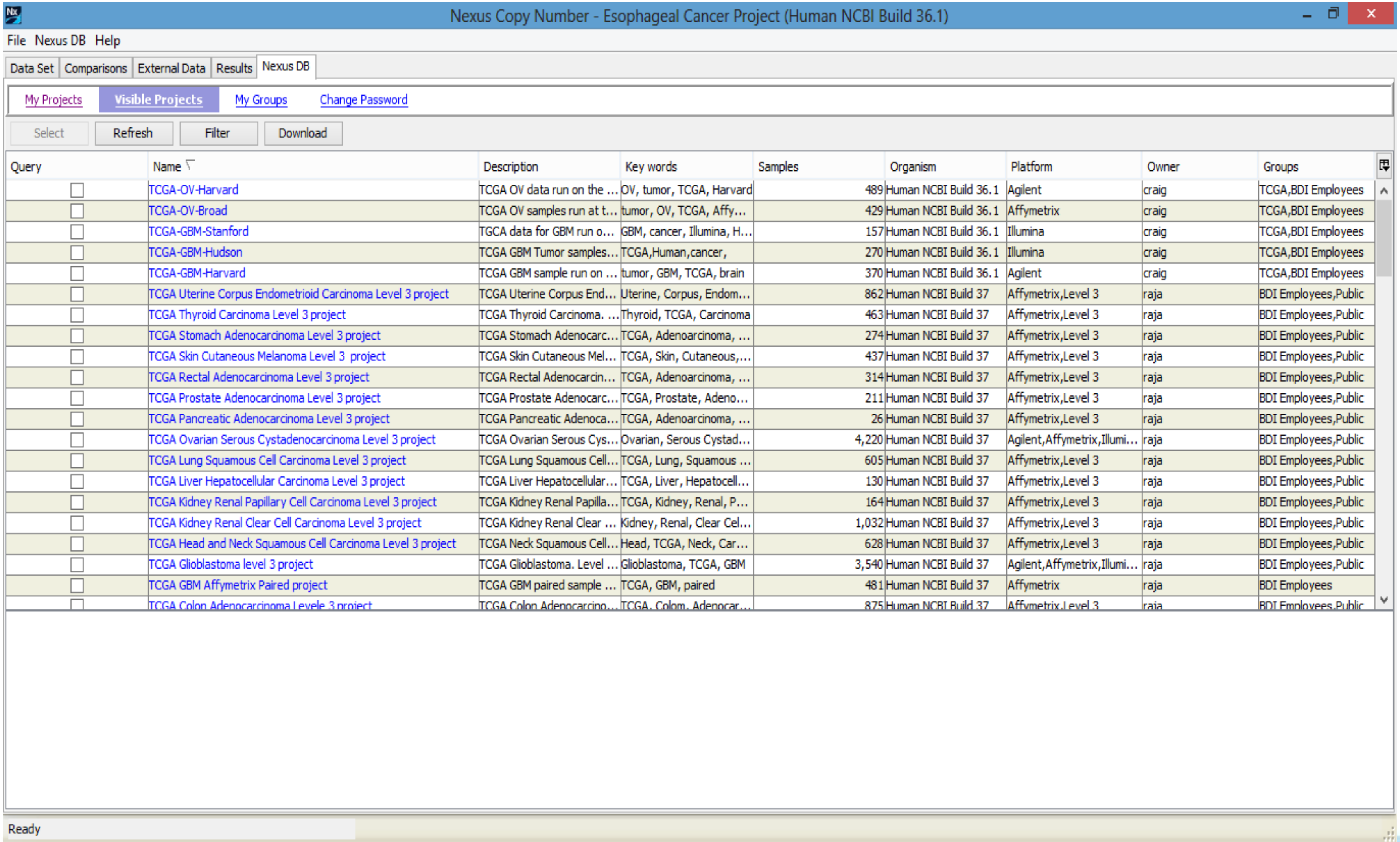
Phone:

User name: tcga_training

Password: TCGAtraining14!

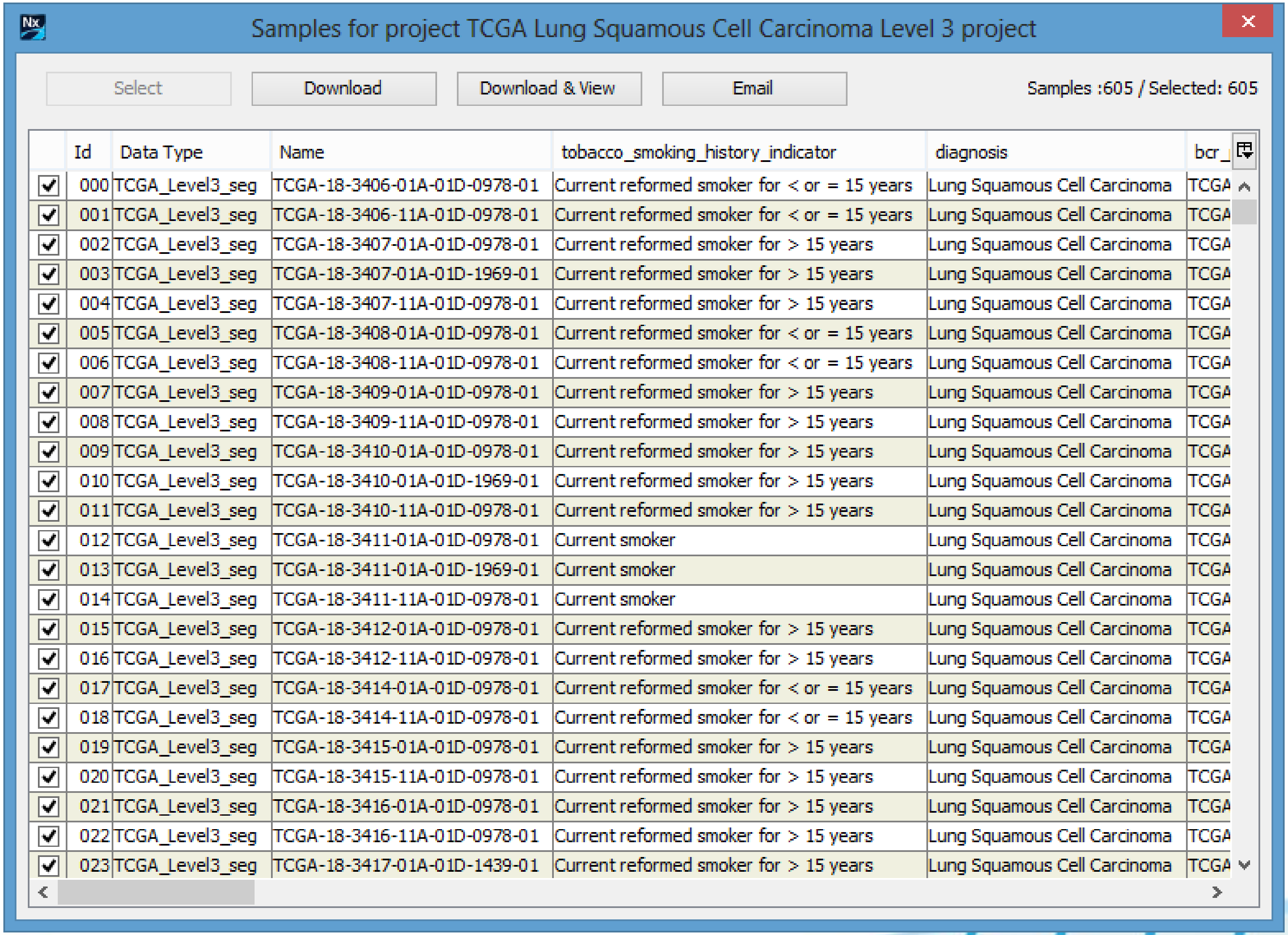
http://chilp.it/0209fe

Nexus DB Data Download



Query	Name	Description	Key words	Samples	Organism	Platform	Owner	Groups
☐	TCGA-OV-Harvard	TCGA OV data run on the …	OV, tumor, TCGA, Harvard	489	Human NCBI Build 36.1	Agilent	craig	TCGA,BDI Employees
☐	TCGA-OV-Broad	TCGA OV samples run at t…	tumor, OV, TCGA, Affy…	429	Human NCBI Build 36.1	Affymetrix	craig	TCGA,BDI Employees
☐	TCGA-GBM-Stanford	TGCA data for GBM run o…	GBM, cancer, Illumina, H…	157	Human NCBI Build 36.1	Illumina	craig	TCGA,BDI Employees
☐	TCGA-GBM-Hudson	TCGA GBM Tumor samples…	TCGA,Human,cancer,	270	Human NCBI Build 36.1	Illumina	craig	TCGA,BDI Employees
☐	TCGA-GBM-Harvard	TCGA GBM sample run on …	tumor, GBM, TCGA, brain	370	Human NCBI Build 36.1	Agilent	craig	TCGA,BDI Employees
☐	TCGA Uterine Corpus Endometrioid Carcinoma Level 3 project	TCGA Uterine Corpus End…	Uterine, Corpus, Endom…	862	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Thyroid Carcinoma Level 3 project	TCGA Thyroid Carcinoma. …	Thyroid, TCGA, Carcinoma	463	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Stomach Adenocarcinoma Level 3 project	TCGA Stomach Adenocarc…	TCGA, Adenoarcinoma, …	274	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Skin Cutaneous Melanoma Level 3 project	TCGA Skin Cutaneous Mel…	TCGA, Skin, Cutaneous,…	437	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Rectal Adenocarcinoma Level 3 project	TCGA Rectal Adenocarcin…	TCGA, Adenoarcinoma, …	314	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Prostate Adenocarcinoma Level 3 project	TCGA Prostate Adenocarc…	TCGA, Prostate, Adeno…	211	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Pancreatic Adenocarcinoma Level 3 project	TCGA Pancreatic Adenoca…	TCGA, Adenoarcinoma, …	26	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Ovarian Serous Cystadenocarcinoma Level 3 project	TCGA Ovarian Serous Cys…	Ovarian, Serous Cystad…	4,220	Human NCBI Build 37	Agilent,Affymetrix,Illumi…	raja	BDI Employees,Public
☐	TCGA Lung Squamous Cell Carcinoma Level 3 project	TCGA Lung Squamous Cell…	TCGA, Lung, Squamous …	605	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Liver Hepatocellular Carcinoma Level 3 project	TCGA Liver Hepatocellular…	TCGA, Liver, Hepatocell…	130	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Kidney Renal Papillary Cell Carcinoma Level 3 project	TCGA Kidney Renal Papilla…	TCGA, Kidney, Renal, P…	164	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Kidney Renal Clear Cell Carcinoma Level 3 project	TCGA Kidney Renal Clear …	Kidney, Renal, Clear Cel…	1,032	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Head and Neck Squamous Cell Carcinoma Level 3 project	TCGA Neck Squamous Cell…	Head, TCGA, Neck, Car…	628	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public
☐	TCGA Glioblastoma level 3 project	TCGA Glioblastoma. Level …	Glioblastoma, TCGA, GBM	3,540	Human NCBI Build 37	Agilent,Affymetrix,Illumi…	raja	BDI Employees,Public
☐	TCGA GBM Affymetrix Paired project	TCGA GBM paired sample …	TCGA, GBM, paired	481	Human NCBI Build 37	Affymetrix	raja	BDI Employees
☐	TCGA Colon Adenocarcinoma Levele 3 project	TCGA Colon Adenocarcino…	TCGA, Colom, Adenocar…	875	Human NCBI Build 37	Affymetrix,Level 3	raja	BDI Employees,Public

Nexus DB Data Download


Samples for project TCGA Lung Squamous Cell Carcinoma Level 3 project
Select
Download
Download & View
Email

Samples :605 / Selected: 605

	Id	Data Type	Name	tobacco_smoking_history_indicator	diagnosis	bcr_
☑	000	TCGA_Level3_seg	TCGA-18-3406-01A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	001	TCGA_Level3_seg	TCGA-18-3406-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	002	TCGA_Level3_seg	TCGA-18-3407-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	003	TCGA_Level3_seg	TCGA-18-3407-01A-01D-1969-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	004	TCGA_Level3_seg	TCGA-18-3407-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	005	TCGA_Level3_seg	TCGA-18-3408-01A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	006	TCGA_Level3_seg	TCGA-18-3408-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	007	TCGA_Level3_seg	TCGA-18-3409-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	008	TCGA_Level3_seg	TCGA-18-3409-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	009	TCGA_Level3_seg	TCGA-18-3410-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	010	TCGA_Level3_seg	TCGA-18-3410-01A-01D-1969-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	011	TCGA_Level3_seg	TCGA-18-3410-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	012	TCGA_Level3_seg	TCGA-18-3411-01A-01D-0978-01	Current smoker	Lung Squamous Cell Carcinoma	TCGA
☑	013	TCGA_Level3_seg	TCGA-18-3411-01A-01D-1969-01	Current smoker	Lung Squamous Cell Carcinoma	TCGA
☑	014	TCGA_Level3_seg	TCGA-18-3411-11A-01D-0978-01	Current smoker	Lung Squamous Cell Carcinoma	TCGA
☑	015	TCGA_Level3_seg	TCGA-18-3412-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	016	TCGA_Level3_seg	TCGA-18-3412-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	017	TCGA_Level3_seg	TCGA-18-3414-01A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	018	TCGA_Level3_seg	TCGA-18-3414-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	019	TCGA_Level3_seg	TCGA-18-3415-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	020	TCGA_Level3_seg	TCGA-18-3415-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	021	TCGA_Level3_seg	TCGA-18-3416-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	022	TCGA_Level3_seg	TCGA-18-3416-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA
☑	023	TCGA_Level3_seg	TCGA-18-3417-01A-01D-1439-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCGA

KICH in The Cancer Genome Atlas (TCGA)

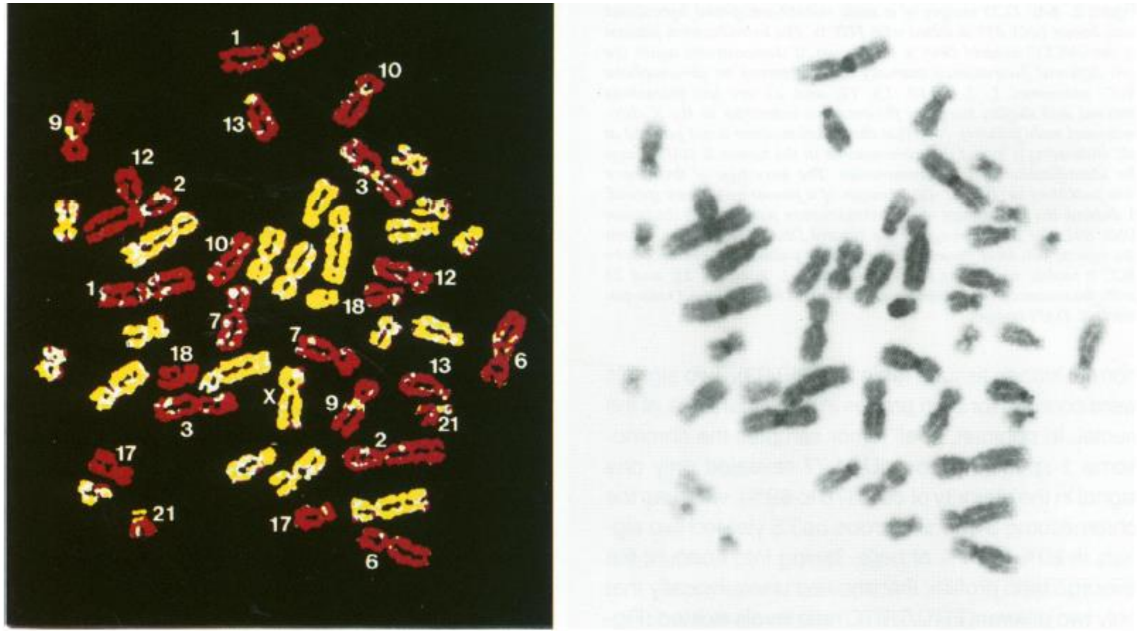
Part of “Rare Tumor” characterization effort.
5% of kidney cancers; Only current therapy is surgery.
TCGA Samples are sporadic (not associated with Birt-Hogg-Dubé syndrome)

What types of discoveries about chromophobe kidney cancer do TCGA researchers hope to make?

- Identify unique patterns of genomic changes that distinguish this subtype of kidney cancer.
- Examine genetic markers that may be predictive of metastasis.
- Determine if a specific genomic signature is connected to tumor recurrence after surgery.
- Genomically differentiate chromophobe kidney cancer and clear cell kidney cancer.
- Gain understanding of the genomic basis of chromophobe kidney cancer to provide the foundation for the development of an effective form of therapy for patients.

KICH Characteristic Whole Chromosome Losses

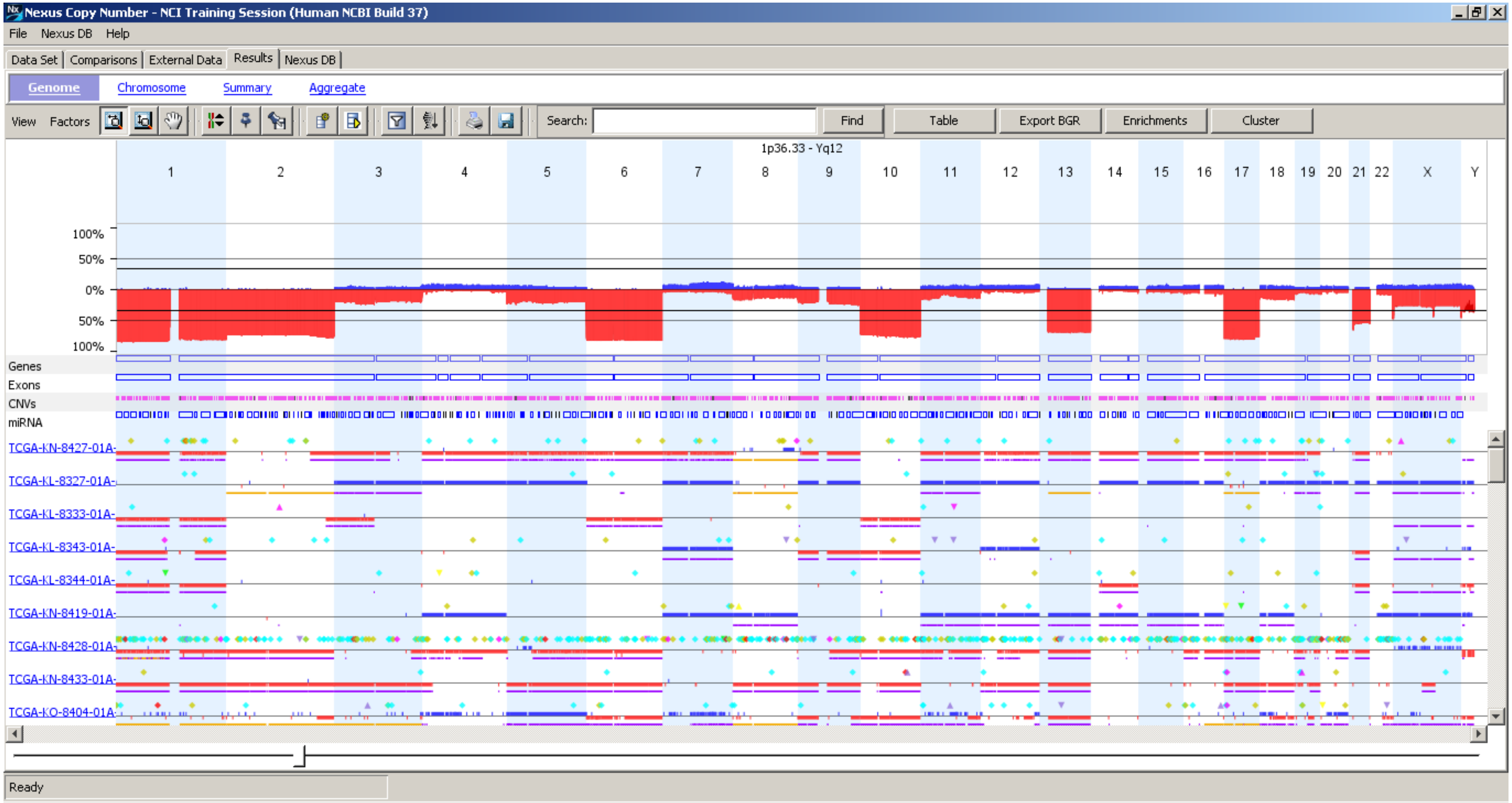
Speicher, M. R., Schoell, B., du Manoir, S., Schröck, E., Ried, T., Cremer, T., ... & Kovacs, G. (1994). **Specific loss of chromosomes 1, 2, 6, 10, 13, 17, and 21 in chromophobe renal cell carcinomas revealed by comparative genomic hybridization.** The American journal of pathology, 145(2), 356.


1
10
9
13
12
2
3
10
12
1
18
7
7
18
13
6
X
9
21
3
2
17
21
17
6

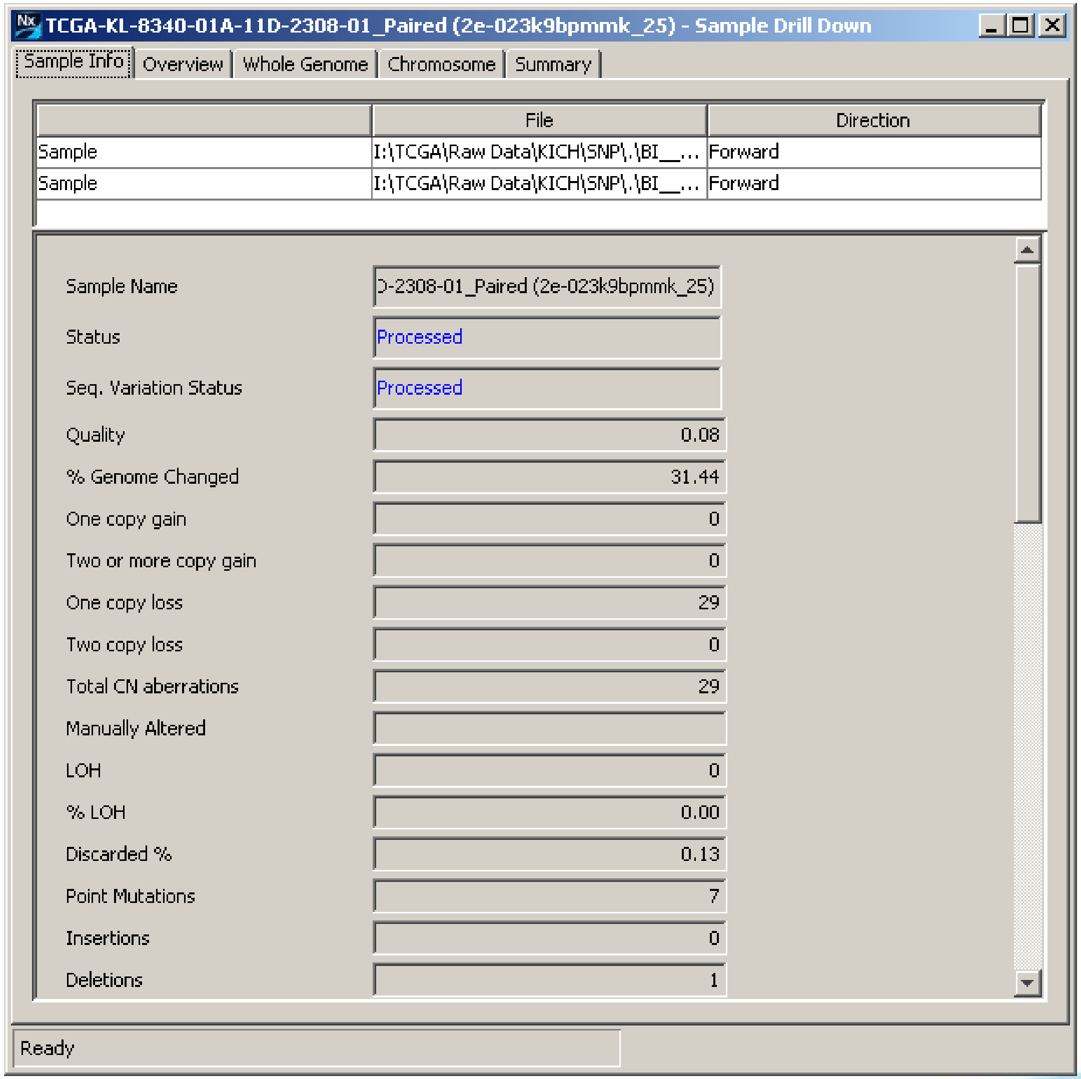
Yellow = balanced between tumor and germline. Red = less DNA in tumor sample.

Downstream Analysis

Results – Genome View


Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File Nexus DB Help
Data Set | Comparisons | External Data | Results | Nexus DB
Genome Chromosome Summary Aggregate
View Factors
Search:
Find
Table
Export BGR
Enrichments
Cluster
1p36.33 - Yq12
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 X Y
100%
50%
0%
50%
100%
Genes
Exons
CNVs
miRNA
TCGA-KN-8427-01A-
TCGA-KL-8327-01A-
TCGA-KL-8333-01A-
TCGA-KL-8343-01A-
TCGA-KL-8344-01A-
TCGA-KN-8419-01A-
TCGA-KN-8428-01A-
TCGA-KN-8433-01A-
TCGA-KO-8404-01A-
Ready

Single Sample Drill Down – Sample Info

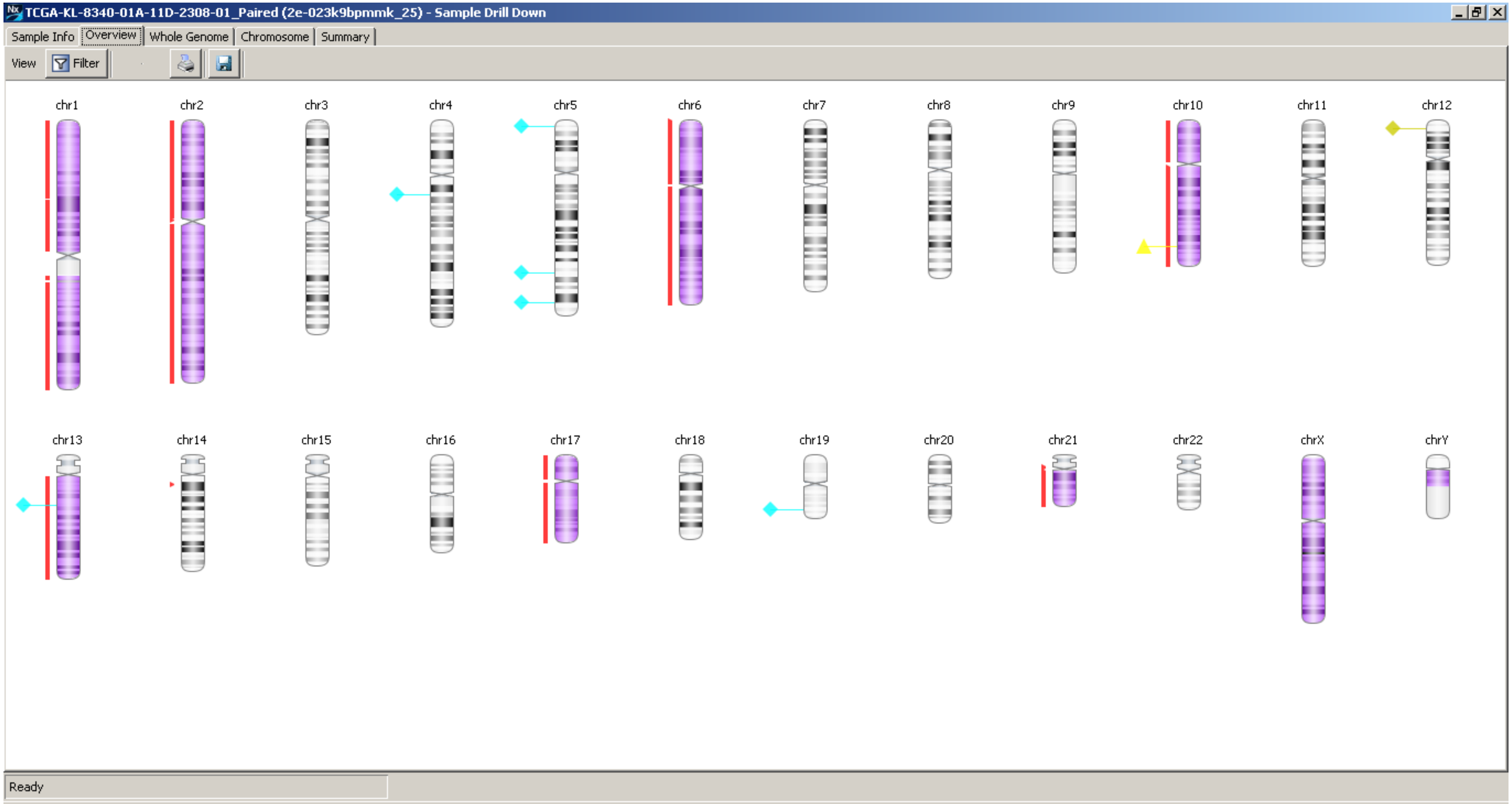

Sample Info
Overview
Whole Genome
Chromosome
Summary
Ready

TCGA-KL-8340-01A-11D-2308-01_Paired (2e-023k9bpmmk_25) - Sample Drill Down

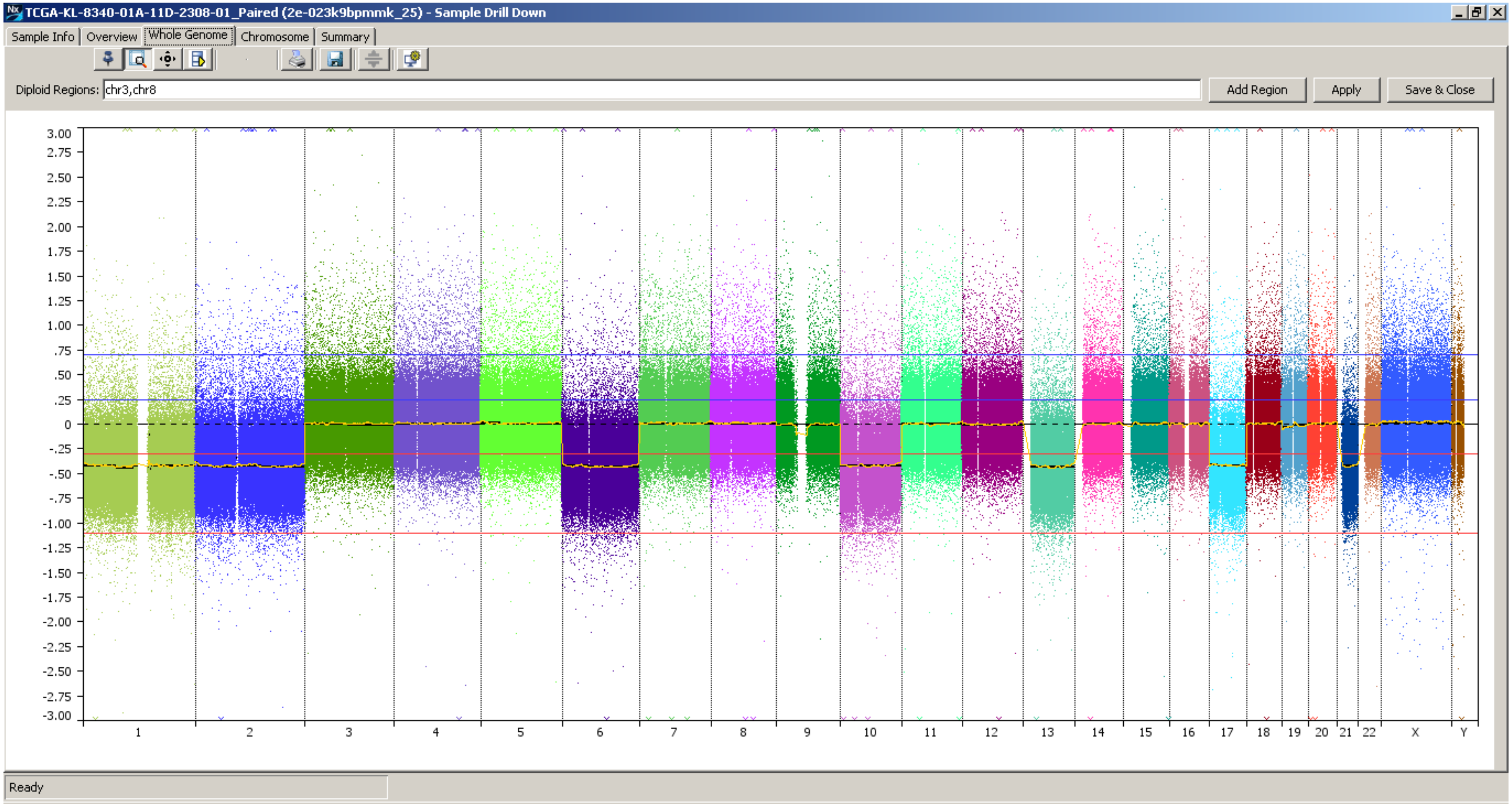
	File	Direction
Sample	I:\TCGA\Raw Data\KICH\SNP\.\BI__...	Forward
Sample	I:\TCGA\Raw Data\KICH\SNP\.\BI__...	Forward

Sample Name	D-2308-01_Paired (2e-023k9bpmmk_25)
Status	Processed
Seq. Variation Status	Processed
Quality	0.08
% Genome Changed	31.44
One copy gain	0
Two or more copy gain	0
One copy loss	29
Two copy loss	0
Total CN aberrations	29
Manually Altered	
LOH	0
% LOH	0.00
Discarded %	0.13
Point Mutations	7
Insertions	0
Deletions	1

Single Sample Drill Down – Overview


TCGA-KL-8340-01A-11D-2308-01_Paired (2e-023k9bpmmk_25) - Sample Drill Down
Sample Info
Overview
Whole Genome
Chromosome
Summary
View
Filter
chr1
chr2
chr3
chr4
chr5
chr6
chr7
chr8
chr9
chr10
chr11
chr12
chr13
chr14
chr15
chr16
chr17
chr18
chr19
chr20
chr21
chr22
chrX
chrY
Ready

Single Sample Drill Down – Whole Genome



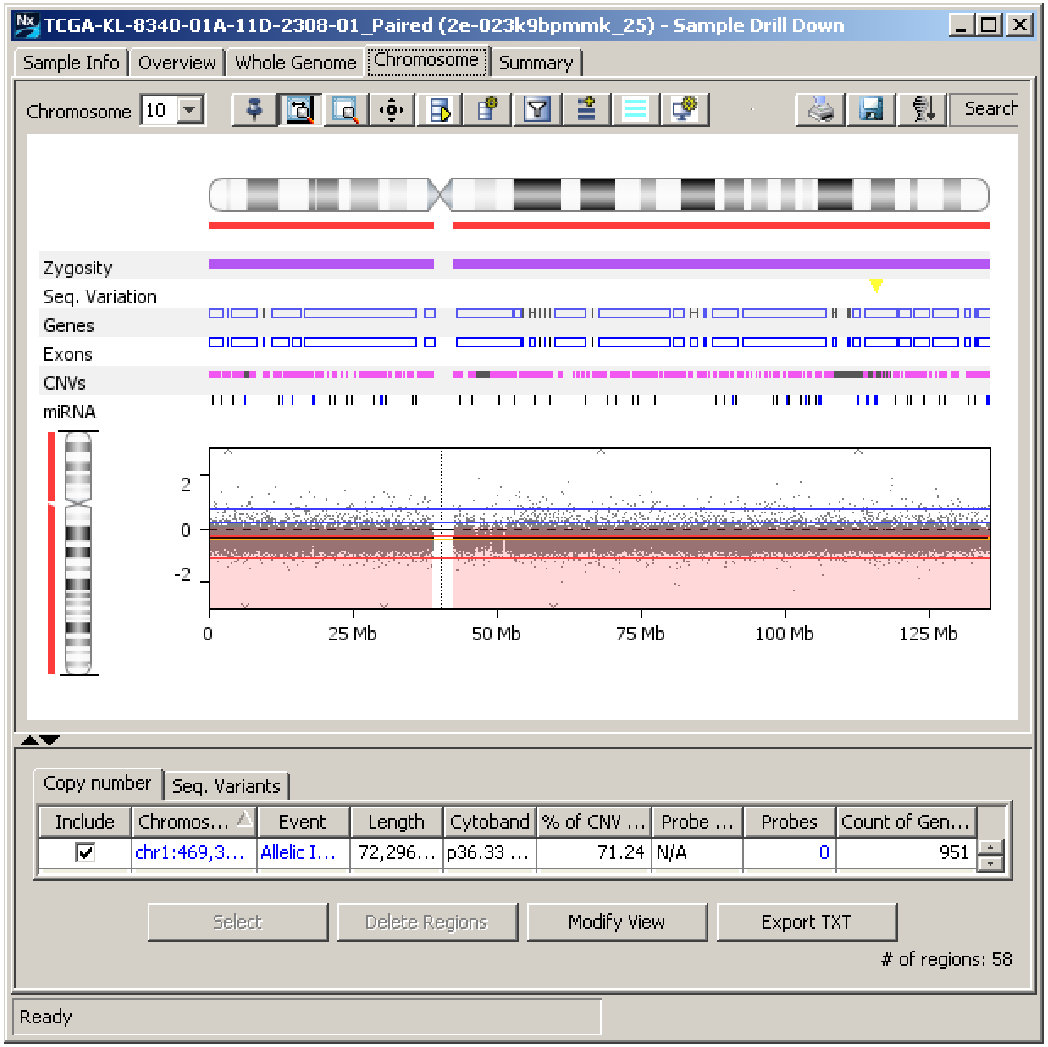
TCGA-KL-8340-01A-11D-2308-01_Paired (2e-023k9bpmmk_25) - Sample Drill Down

Diploid Regions: chr3,chr8

Ready

Sample Info
Overview
Whole Genome
Chromosome
Summary
Add Region
Apply
Save & Close
3.00
2.75
2.50
2.25
2.00
1.75
1.50
1.25
1.00
.75
.50
.25
0
-.25
-.50
-.75
-1.00
-1.25
-1.50
-1.75
-2.00
-2.25
-2.50
-2.75
-3.00
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y

Single Sample Drill Down – Chromosome


TCGA-KL-8340-01A-11D-2308-01_Paired (2e-023k9bpmmk_25) - Sample Drill Down

Chromosome 10

Include	Chromos...	Event	Length	Cytoband	% of CNV ...	Probe ...	Probes	Count of Gen...
☑	chr1:469,3...	Allelic I...	72,296...	p36.33 ...	71.24	N/A	0	951

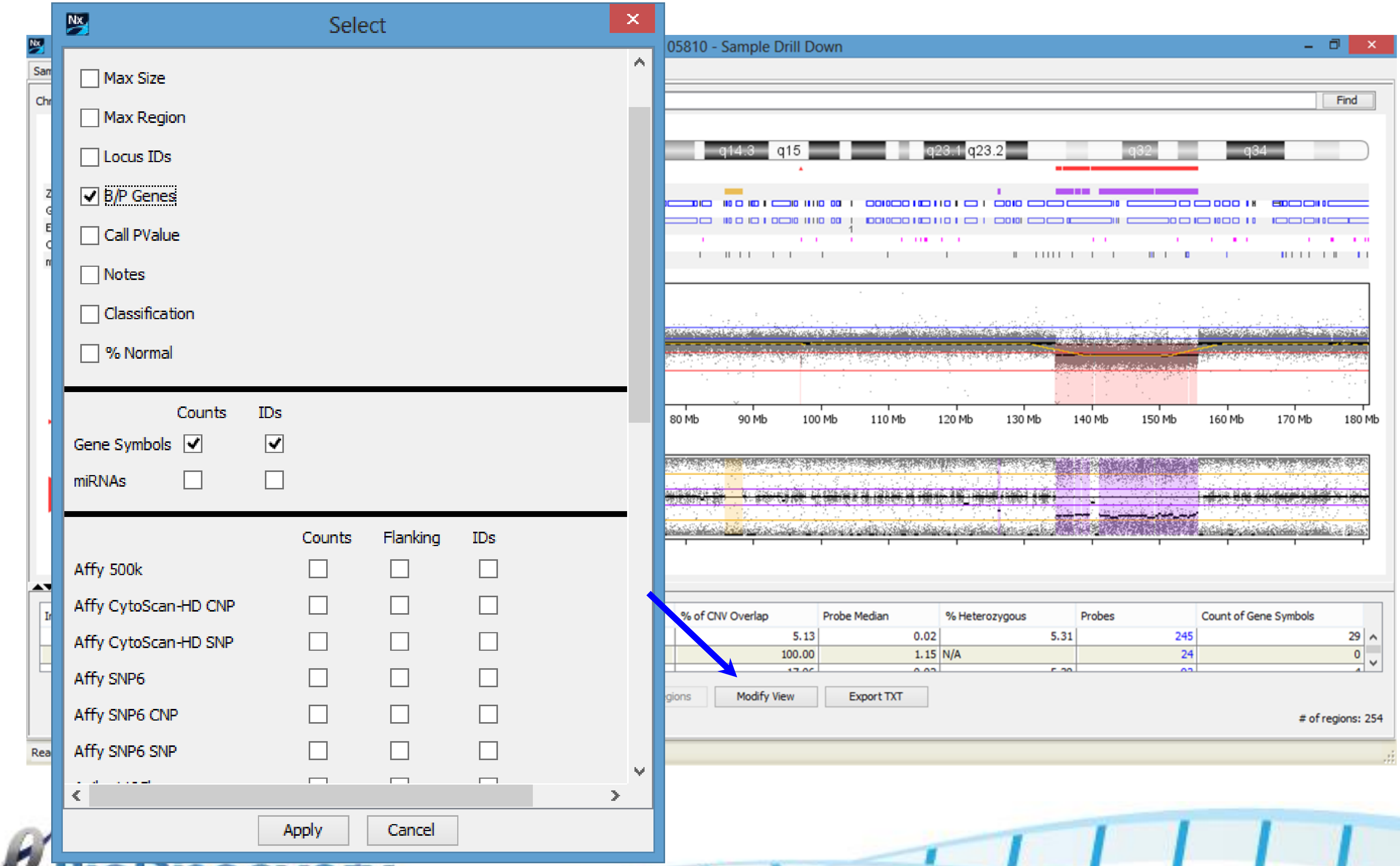
# of regions: 58

Ready

Sample Info
Overview
Whole Genome
Chromosome
Summary
Search
Zygosity
Seq. Variation
Genes
Exons
CNVs
miRNA
2
0
-2
0
25 Mb
50 Mb
75 Mb
100 Mb
125 Mb
Copy number
Seq. Variants
Select
Delete Regions
Modify View
Export TXT

- Mark Location
- Horizontal Zoom
- Rectangular Zoom
- Center View
- Locate Region
- Query Database
- Region Query Settings
- Filter
- Add/Join Region
- Modify Tracks

Single Sample Drill Down – Modify View



Select
05810 - Sample Drill Down
Find
q14.3
q15
q23.1
q23.2
q32
q34
80 Mb
90 Mb
100 Mb
110 Mb
120 Mb
130 Mb
140 Mb
150 Mb
160 Mb
170 Mb
180 Mb
gions
Modify View
Export TXT
Apply
Cancel

☐ Max Size
☐ Max Region
☐ Locus IDs
☑ B/P Genes
☐ Call PValue
☐ Notes
☐ Classification
☐ % Normal

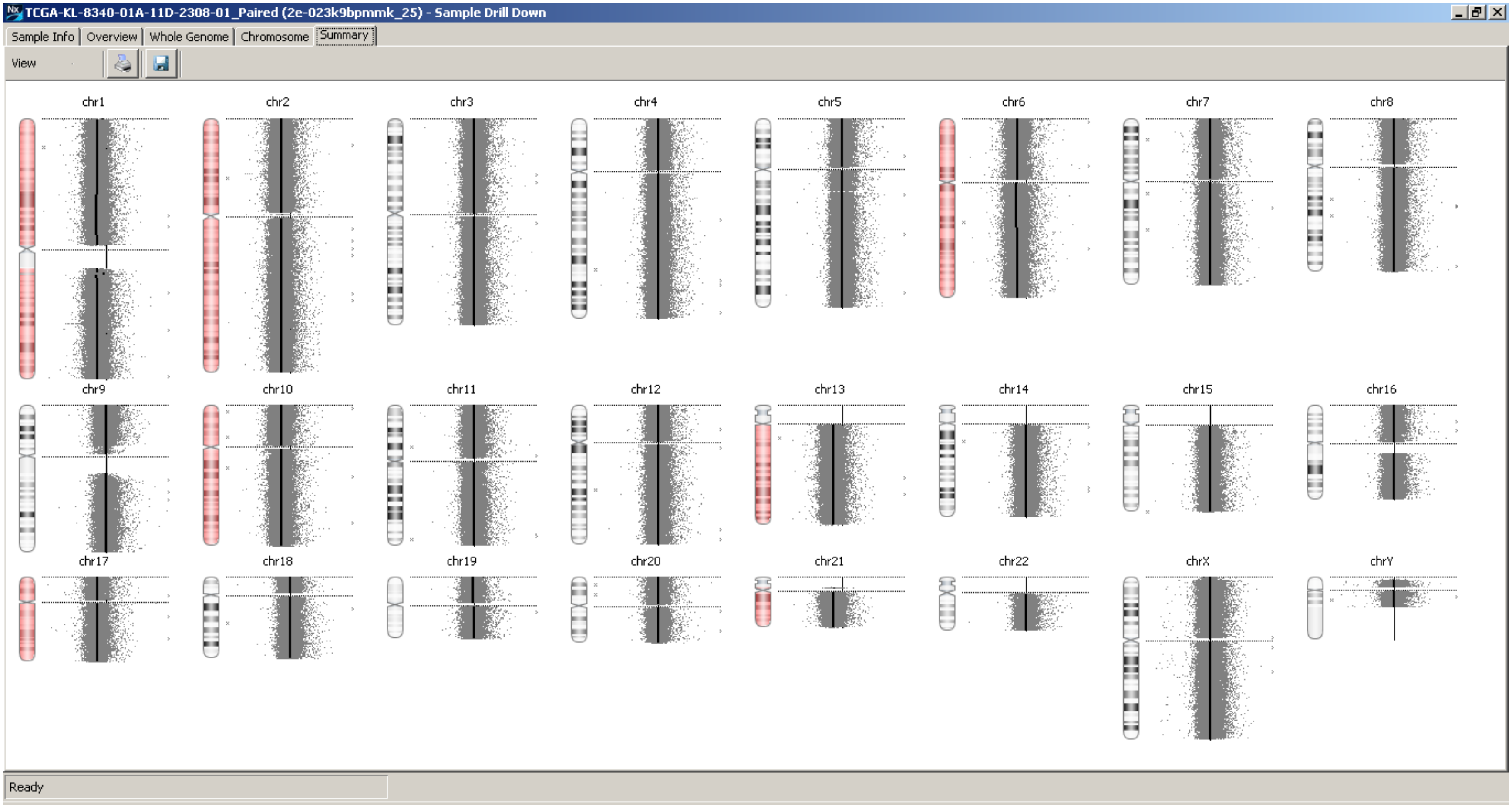
	Counts	IDs
Gene Symbols	☑	☑
miRNAs	☐	☐

	Counts	Flanking	IDs
Affy 500k	☐	☐	☐
Affy CytoScan-HD CNP	☐	☐	☐
Affy CytoScan-HD SNP	☐	☐	☐
Affy SNP6	☐	☐	☐
Affy SNP6 CNP	☐	☐	☐
Affy SNP6 SNP	☐	☐	☐

% of CNV Overlap	Probe Median	% Heterozygous	Probes	Count of Gene Symbols
5.13	0.02	5.31	245	29
100.00	1.15	N/A	24	0

# of regions: 254

Single Sample Drill Down – Summary


TCGA-KL-8340-01A-11D-2308-01_Paired (2e-023k9bpmmk_25) - Sample Drill Down
Sample Info
Overview
Whole Genome
Chromosome
Summary
View
chr1
chr2
chr3
chr4
chr5
chr6
chr7
chr8
chr9
chr10
chr11
chr12
chr13
chr14
chr15
chr16
chr17
chr18
chr19
chr20
chr21
chr22
chrX
chrY
Ready

Identifying Frequent and Significant Peaks

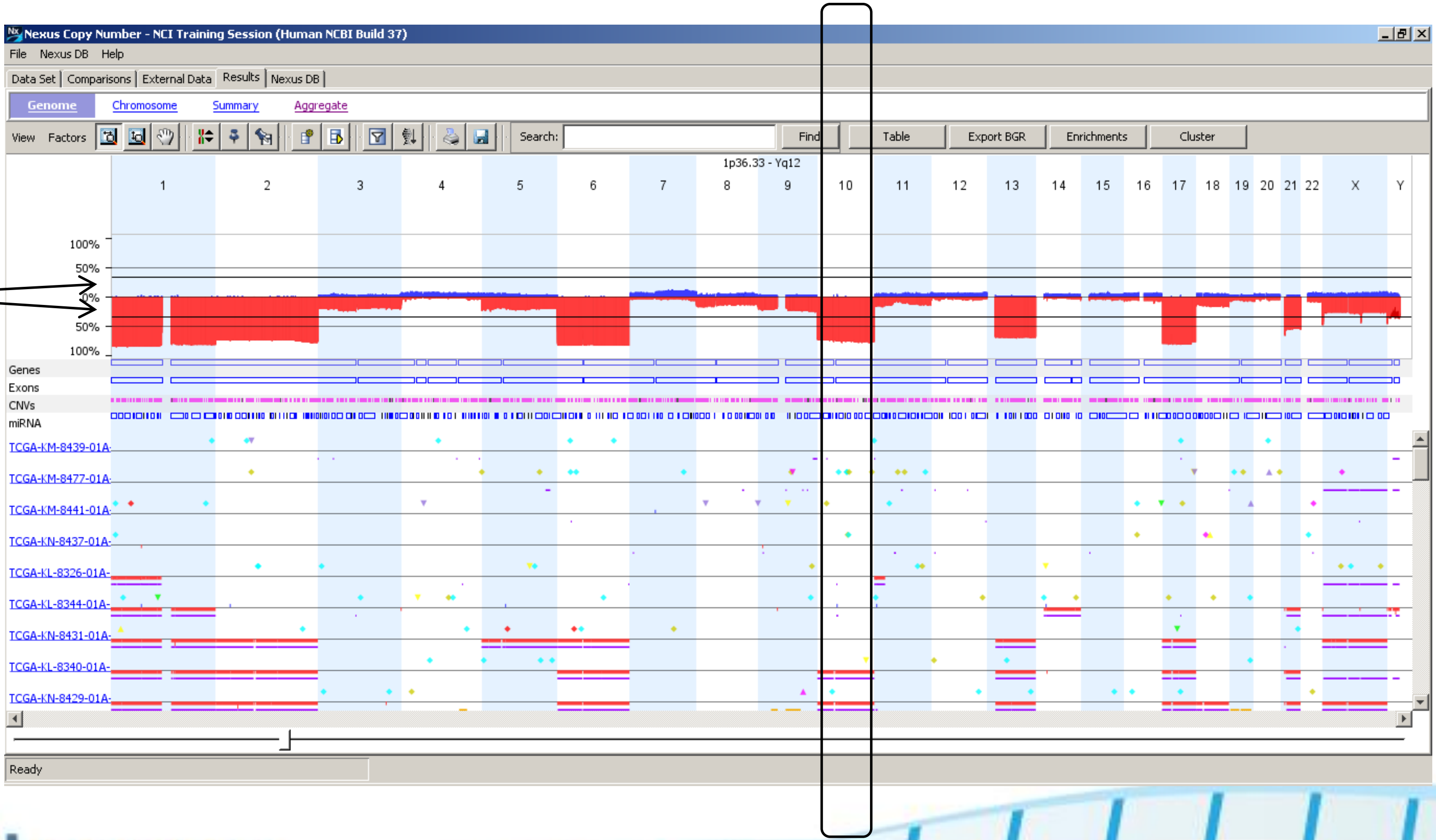
- What are “common” aberrations in this set
- What are “significant” common abberrations

Aggregate Table Approach

- Find regions that have an aberration in at least x% of the samples

Peak

35%



Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Results
Nexus DB
Genome
Chromosome
Summary
Aggregate
View
Factors
Search:
Find
Table
Export BGR
Enrichments
Cluster
1p36.33 - Yq12
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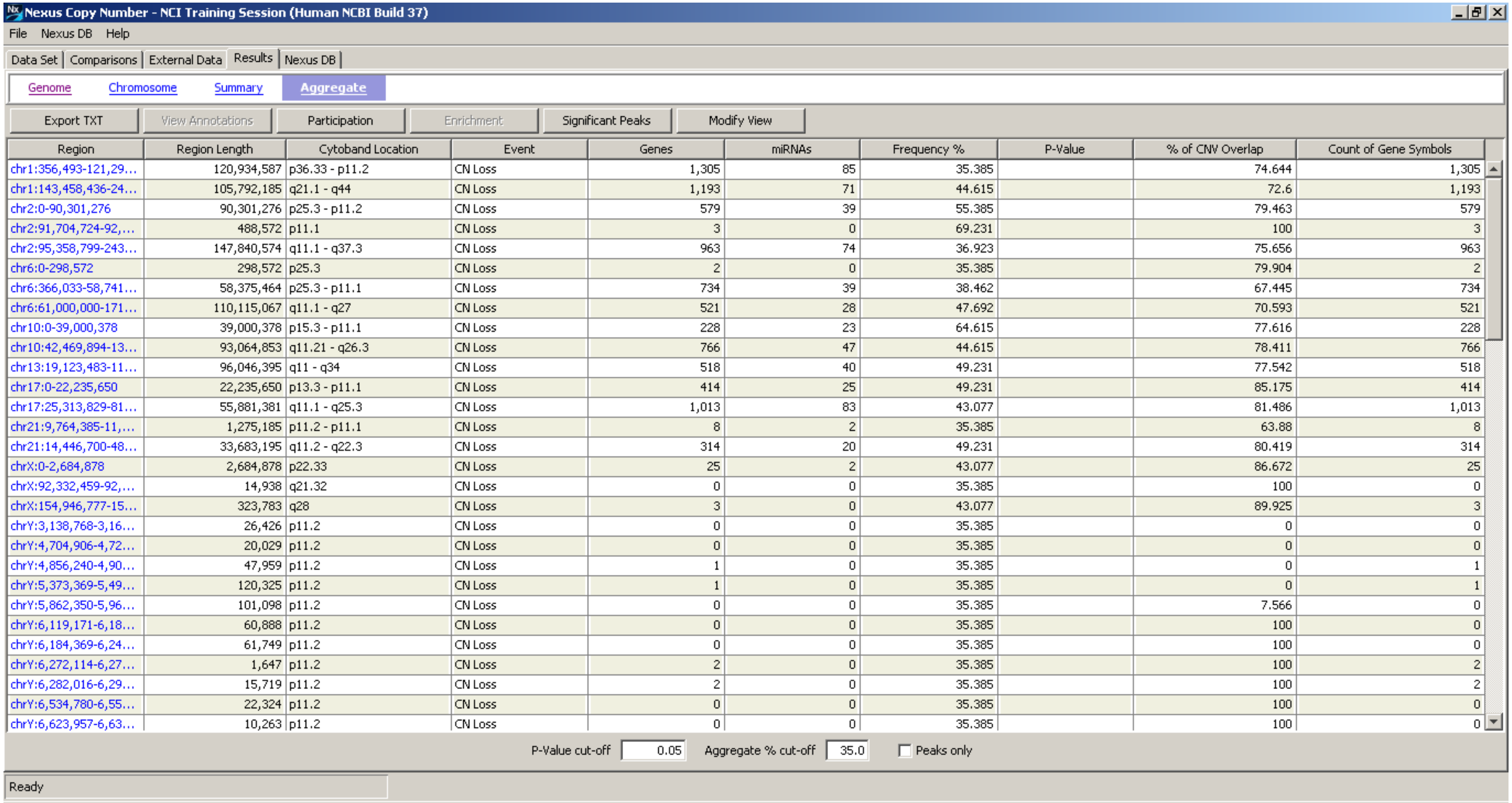
Genes
Exons
CNVs
miRNA
TCGA-KM-8439-01A-
TCGA-KM-8477-01A-
TCGA-KM-8441-01A-
TCGA-KN-8437-01A-
TCGA-KL-8326-01A-
TCGA-KL-8344-01A-
TCGA-KN-8431-01A-
TCGA-KL-8340-01A-
TCGA-KN-8429-01A-
Ready

Aggregate Table


Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File
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Data Set
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External Data
Results
Nexus DB
Genome
Chromosome
Summary
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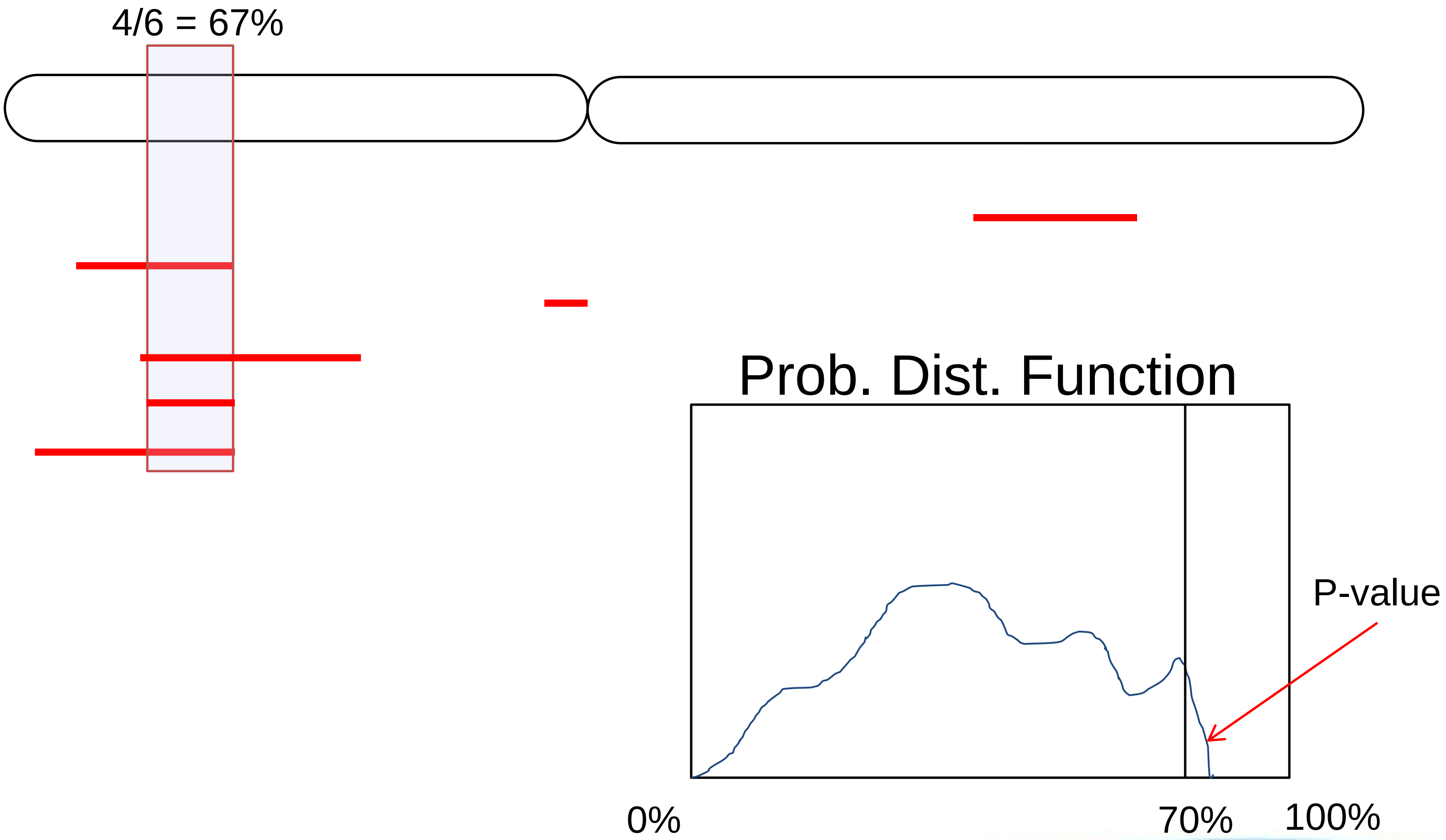
Export TXT
View Annotations
Participation
Enrichment
Significant Peaks
Modify View

Region	Region Length	Cytoband Location	Event	Genes	miRNAs	Frequency %	P-Value	% of CNV Overlap	Count of Gene Symbols
chr1:356,493-121,29...	120,934,587	p36.33 - p11.2	CN Loss	1,305	85	35.385		74.644	1,305
chr1:143,458,436-24...	105,792,185	q21.1 - q44	CN Loss	1,193	71	44.615		72.6	1,193
chr2:0-90,301,276	90,301,276	p25.3 - p11.2	CN Loss	579	39	55.385		79.463	579
chr2:91,704,724-92,...	488,572	p11.1	CN Loss	3	0	69.231		100	3
chr2:95,358,799-243...	147,840,574	q11.1 - q37.3	CN Loss	963	74	36.923		75.656	963
chr6:0-298,572	298,572	p25.3	CN Loss	2	0	35.385		79.904	2
chr6:366,033-58,741...	58,375,464	p25.3 - p11.1	CN Loss	734	39	38.462		67.445	734
chr6:61,000,000-171...	110,115,067	q11.1 - q27	CN Loss	521	28	47.692		70.593	521
chr10:0-39,000,378	39,000,378	p15.3 - p11.1	CN Loss	228	23	64.615		77.616	228
chr10:42,469,894-13...	93,064,853	q11.21 - q26.3	CN Loss	766	47	44.615		78.411	766
chr13:19,123,483-11...	96,046,395	q11 - q34	CN Loss	518	40	49.231		77.542	518
chr17:0-22,235,650	22,235,650	p13.3 - p11.1	CN Loss	414	25	49.231		85.175	414
chr17:25,313,829-81...	55,881,381	q11.1 - q25.3	CN Loss	1,013	83	43.077		81.486	1,013
chr21:9,764,385-11,...	1,275,185	p11.2 - p11.1	CN Loss	8	2	35.385		63.88	8
chr21:14,446,700-48...	33,683,195	q11.2 - q22.3	CN Loss	314	20	49.231		80.419	314
chrX:0-2,684,878	2,684,878	p22.33	CN Loss	25	2	43.077		86.672	25
chrX:92,332,459-92,...	14,938	q21.32	CN Loss	0	0	35.385		100	0
chrX:154,946,777-15...	323,783	q28	CN Loss	3	0	43.077		89.925	3
chrY:3,138,768-3,16...	26,426	p11.2	CN Loss	0	0	35.385		0	0
chrY:4,704,906-4,72...	20,029	p11.2	CN Loss	0	0	35.385		0	0
chrY:4,856,240-4,90...	47,959	p11.2	CN Loss	1	0	35.385		0	1
chrY:5,373,369-5,49...	120,325	p11.2	CN Loss	1	0	35.385		0	1
chrY:5,862,350-5,96...	101,098	p11.2	CN Loss	0	0	35.385		7.566	0
chrY:6,119,171-6,18...	60,888	p11.2	CN Loss	0	0	35.385		100	0
chrY:6,184,369-6,24...	61,749	p11.2	CN Loss	0	0	35.385		100	0
chrY:6,272,114-6,27...	1,647	p11.2	CN Loss	2	0	35.385		100	2
chrY:6,282,016-6,29...	15,719	p11.2	CN Loss	2	0	35.385		100	2
chrY:6,534,780-6,55...	22,324	p11.2	CN Loss	0	0	35.385		100	0
chrY:6,623,957-6,63...	10,263	p11.2	CN Loss	0	0	35.385		100	0

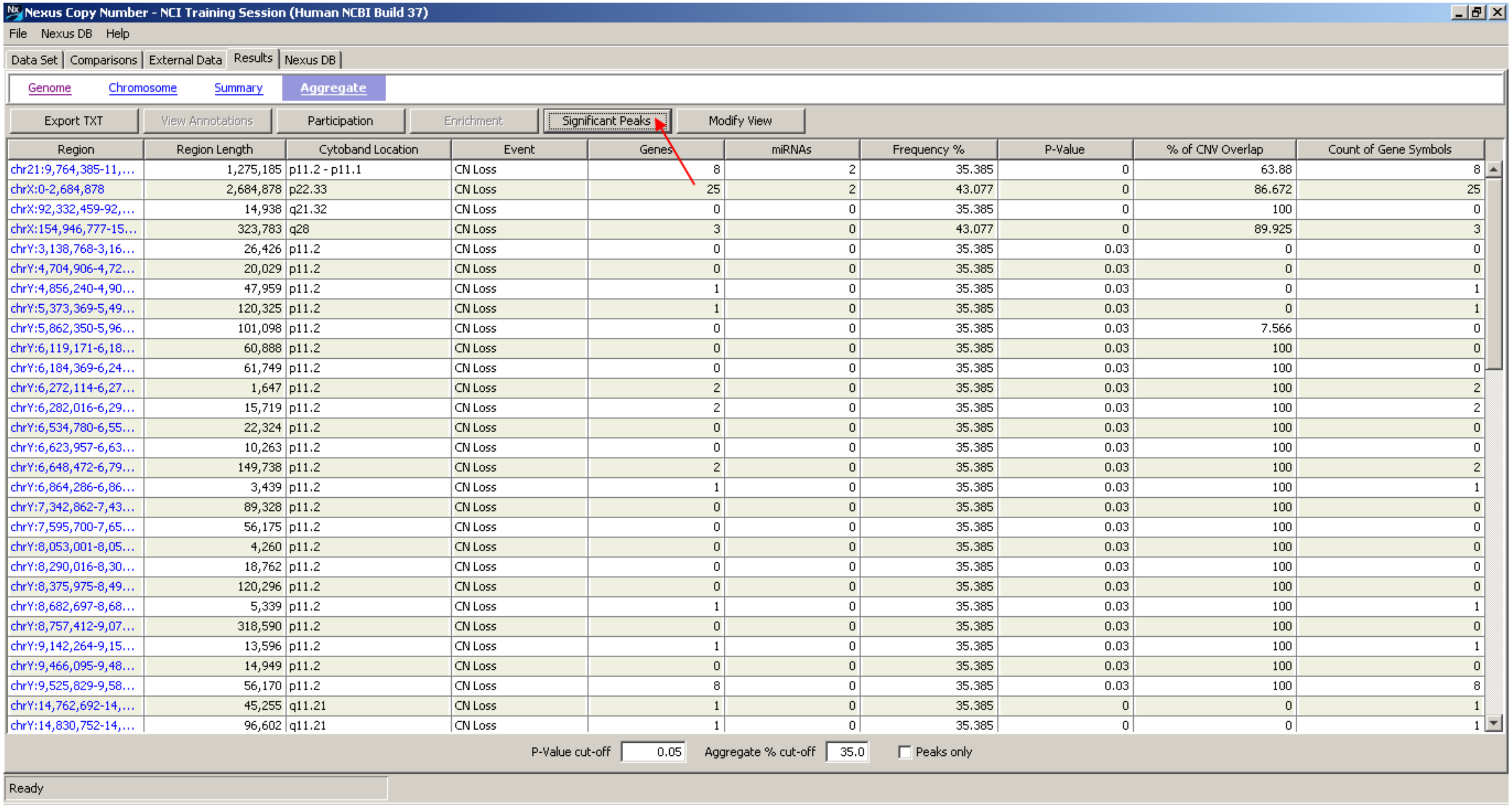
P-Value cut-off 0.05 Aggregate % cut-off 35.0 ☐ Peaks only

Ready

Significance Testing (STAC)


4/6 = 67%
Prob. Dist. Function
P-value
0%
70%
100%

Significant Peaks (STAC)


Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File
Nexus DB
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Genome
Chromosome
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Enrichment
Significant Peaks
Modify View
Ready

Region	Region Length	Cytoband Location	Event	Genes	miRNAs	Frequency %	P-Value	% of CNV Overlap	Count of Gene Symbols
chr21:9,764,385-11,...	1,275,185	p11.2 - p11.1	CN Loss	8	2	35.385	0	63.88	8
chrX:0-2,684,878	2,684,878	p22.33	CN Loss	25	2	43.077	0	86.672	25
chrX:92,332,459-92,...	14,938	q21.32	CN Loss	0	0	35.385	0	100	0
chrX:154,946,777-15...	323,783	q28	CN Loss	3	0	43.077	0	89.925	3
chrY:3,138,768-3,16...	26,426	p11.2	CN Loss	0	0	35.385	0.03	0	0
chrY:4,704,906-4,72...	20,029	p11.2	CN Loss	0	0	35.385	0.03	0	0
chrY:4,856,240-4,90...	47,959	p11.2	CN Loss	1	0	35.385	0.03	0	1
chrY:5,373,369-5,49...	120,325	p11.2	CN Loss	1	0	35.385	0.03	0	1
chrY:5,862,350-5,96...	101,098	p11.2	CN Loss	0	0	35.385	0.03	7.566	0
chrY:6,119,171-6,18...	60,888	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:6,184,369-6,24...	61,749	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:6,272,114-6,27...	1,647	p11.2	CN Loss	2	0	35.385	0.03	100	2
chrY:6,282,016-6,29...	15,719	p11.2	CN Loss	2	0	35.385	0.03	100	2
chrY:6,534,780-6,55...	22,324	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:6,623,957-6,63...	10,263	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:6,648,472-6,79...	149,738	p11.2	CN Loss	2	0	35.385	0.03	100	2
chrY:6,864,286-6,86...	3,439	p11.2	CN Loss	1	0	35.385	0.03	100	1
chrY:7,342,862-7,43...	89,328	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:7,595,700-7,65...	56,175	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:8,053,001-8,05...	4,260	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:8,290,016-8,30...	18,762	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:8,375,975-8,49...	120,296	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:8,682,697-8,68...	5,339	p11.2	CN Loss	1	0	35.385	0.03	100	1
chrY:8,757,412-9,07...	318,590	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:9,142,264-9,15...	13,596	p11.2	CN Loss	1	0	35.385	0.03	100	1
chrY:9,466,095-9,48...	14,949	p11.2	CN Loss	0	0	35.385	0.03	100	0
chrY:9,525,829-9,58...	56,170	p11.2	CN Loss	8	0	35.385	0.03	100	8
chrY:14,762,692-14,...	45,255	q11.21	CN Loss	1	0	35.385	0	0	1
chrY:14,830,752-14,...	96,602	q11.21	CN Loss	1	0	35.385	0	0	1

P-Value cut-off 0.05 Aggregate % cut-off 35.0 ☐ Peaks only

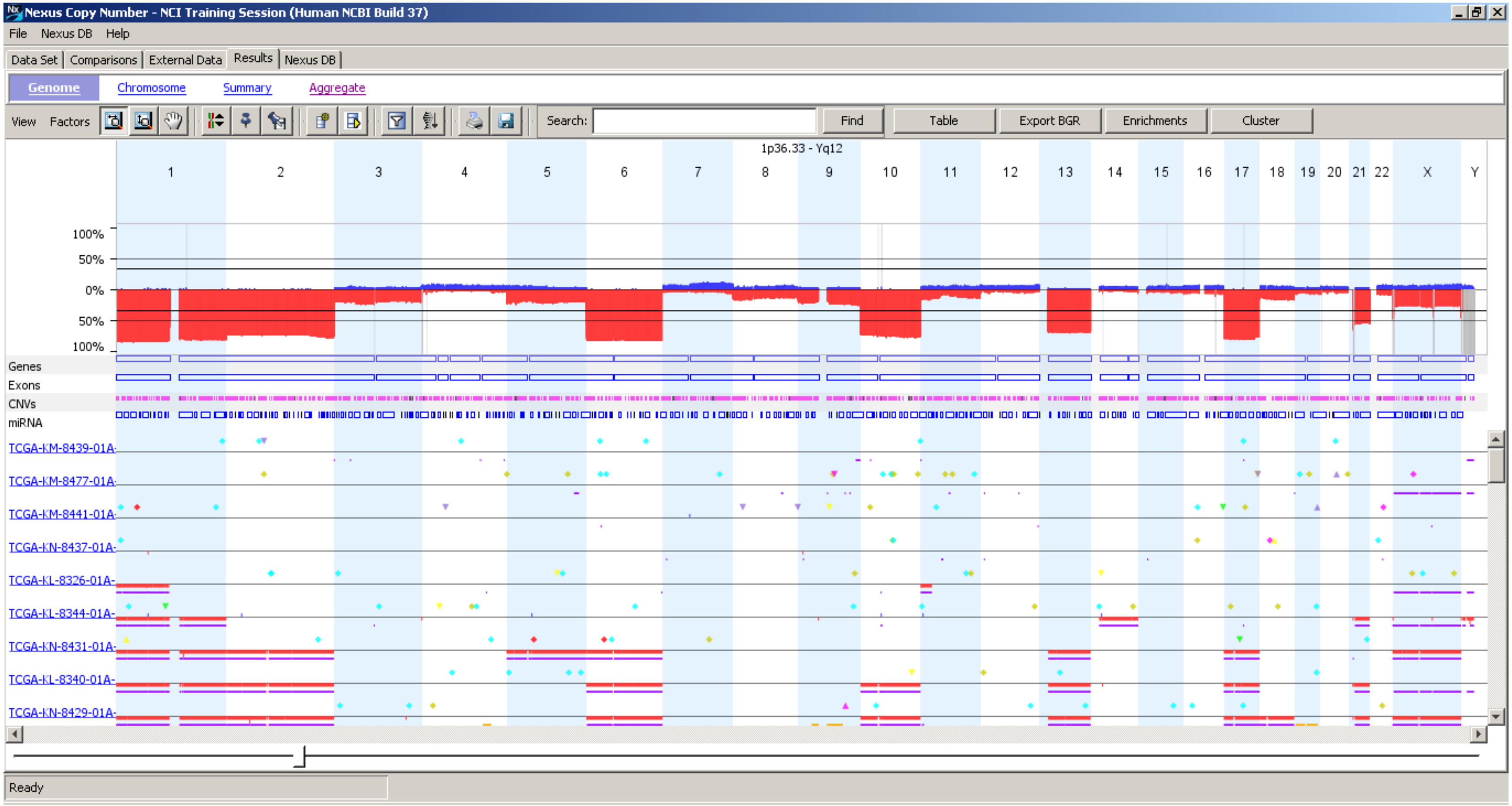
STAC

- Makes all calculations based on current calls
  - Excludes any calls you have removed
  - Includes manually added/adjusted calls
  - Includes gender correction (if applied)

- Will only indicate Peaks
  - Does not include longer extended regions

- Does not take into consideration amplitude
  - High copy gain and single copy gain / homozygous deletion and single copy loss weighted equally

Significant Peaks (STAC)


Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File
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Genes
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TCGA-KM-8439-01A
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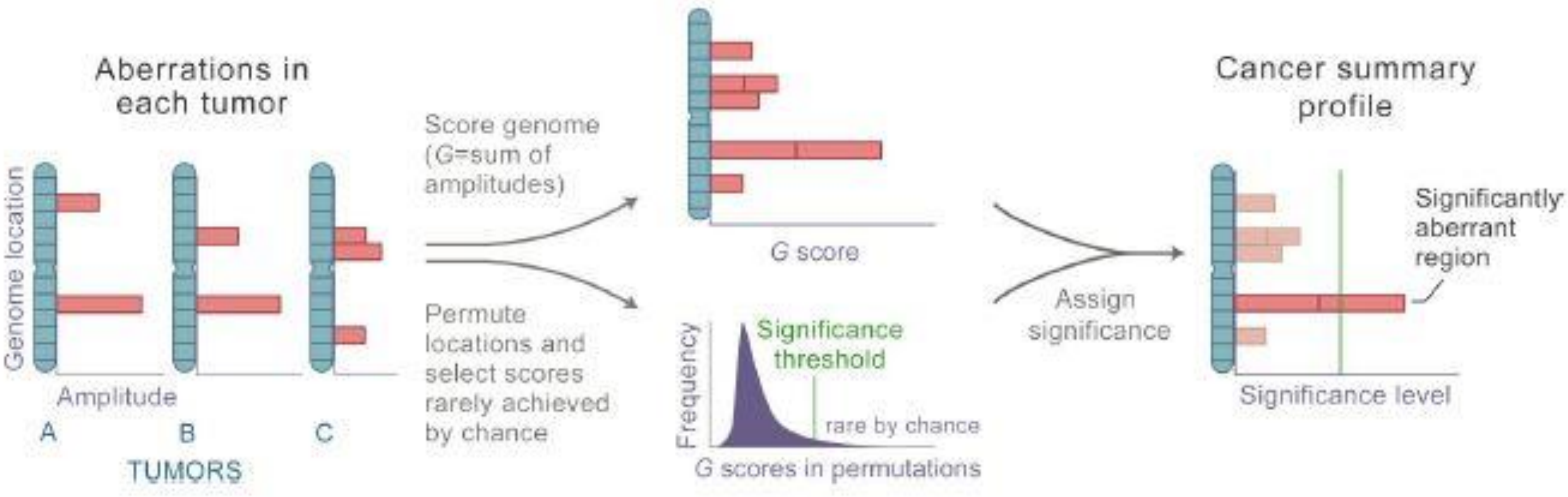
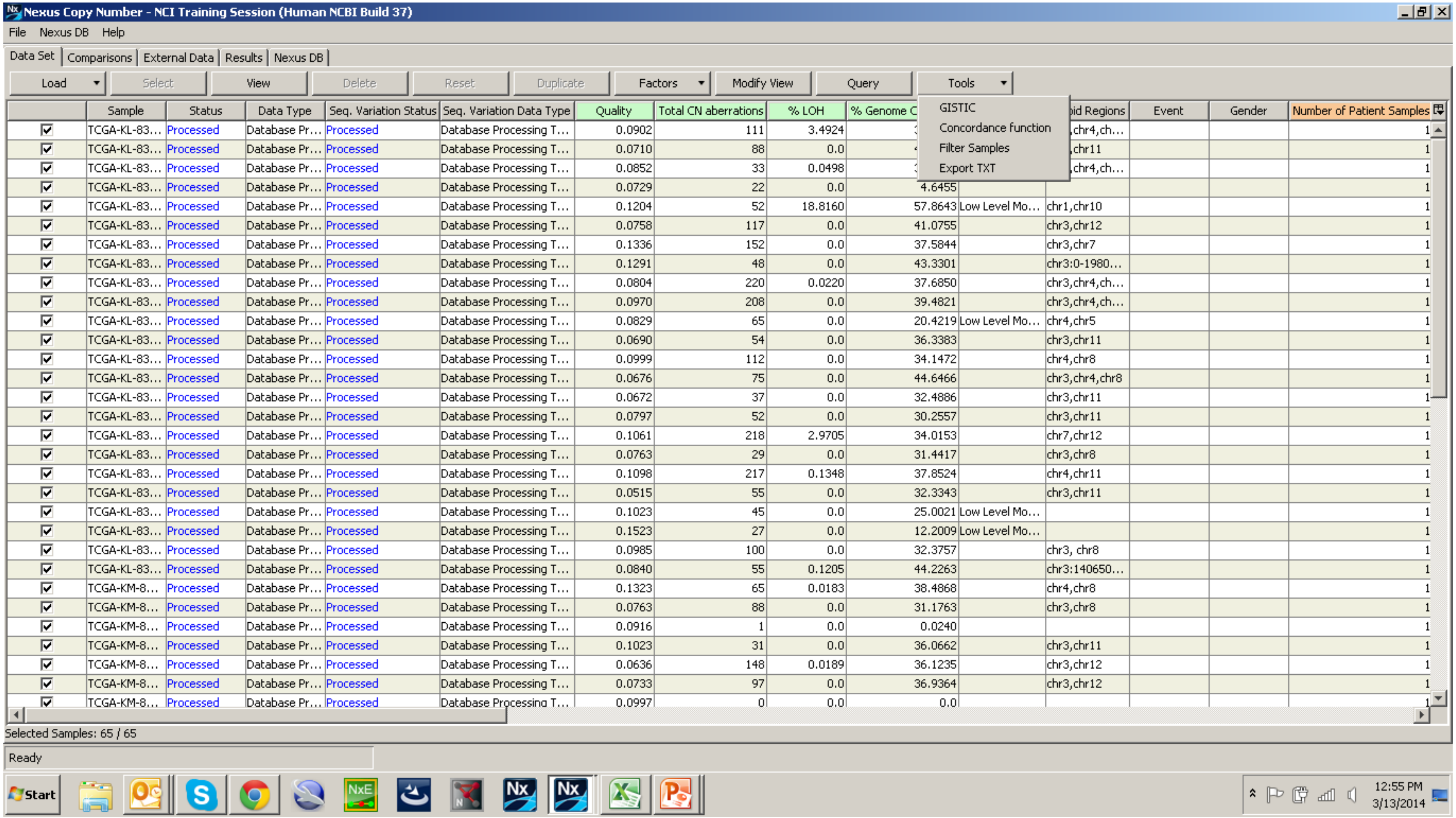
TCGA-KM-8441-01A
TCGA-KN-8437-01A
TCGA-KL-8326-01A
TCGA-KL-8344-01A
TCGA-KN-8431-01A
TCGA-KL-8340-01A
TCGA-KN-8429-01A
Ready

GISTIC


Aberrations in each tumor
Genome location
Amplitude
A
B
C
TUMORS
Score genome (G=sum of amplitudes)
Permute locations and select scores rarely achieved by chance
G score
Frequency
Significance threshold
rare by chance
G scores in permutations
Assign significance
Cancer summary profile
Significantly aberrant region
Significance level

- Specify G-score (combination score of frequency and amplitude) and Q-bound (significance)
  - Only multiple testing correction value calculated, may need to reduce threshold to best capture results (0.25)

GISTIC



	Sample	Status	Data Type	Seq. Variation Status	Seq. Variation Data Type	Quality	Total CN aberrations	% LOH	% Genome C		id Regions	Event	Gender	Number of Patient Samples
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0902	111	3.4924			,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0710	88	0.0			,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0852	33	0.0498			,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0729	22	0.0	4.6455					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1204	52	18.8160	57.8643	Low Level Mo…	chr1,chr10			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0758	117	0.0	41.0755		chr3,chr12			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1336	152	0.0	37.5844		chr3,chr7			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1291	48	0.0	43.3301		chr3:0-1980…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0804	220	0.0220	37.6850		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0970	208	0.0	39.4821		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0829	65	0.0	20.4219	Low Level Mo…	chr4,chr5			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0690	54	0.0	36.3383		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0999	112	0.0	34.1472		chr4,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0676	75	0.0	44.6466		chr3,chr4,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0672	37	0.0	32.4886		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0797	52	0.0	30.2557		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1061	218	2.9705	34.0153		chr7,chr12			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0763	29	0.0	31.4417		chr3,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1098	217	0.1348	37.8524		chr4,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0515	55	0.0	32.3343		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1023	45	0.0	25.0021	Low Level Mo…				1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1523	27	0.0	12.2009	Low Level Mo…				1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0985	100	0.0	32.3757		chr3, chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0840	55	0.1205	44.2263		chr3:140650…			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.1323	65	0.0183	38.4868		chr4,chr8			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0763	88	0.0	31.1763		chr3,chr8			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0916	1	0.0	0.0240					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.1023	31	0.0	36.0662		chr3,chr11			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0636	148	0.0189	36.1235		chr3,chr12			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0733	97	0.0	36.9364		chr3,chr12			1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0997	0	0.0	0.0					1

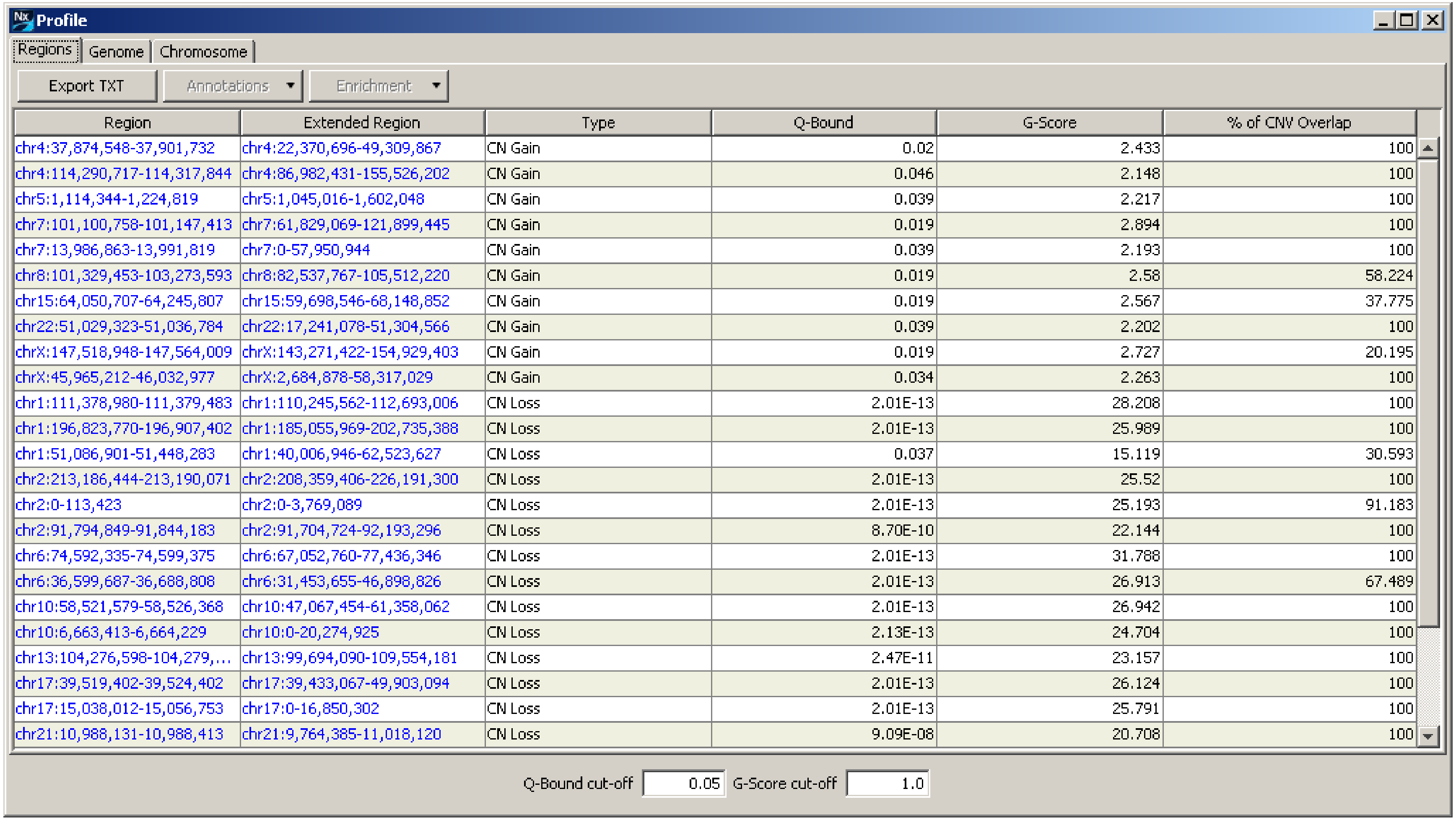
Selected Samples: 65 / 65

Ready

GISTIC

- Indicates Peaks and extended regions
  - Extended regions may include full chromosome
- Makes all calculations based on raw probe values
  - Excludes manual adjustments and gender correction, so sex chromosome calls may be incorrect
- Considers both amplitude and frequency
  - Added weight for high copy gains and homozygous deletions

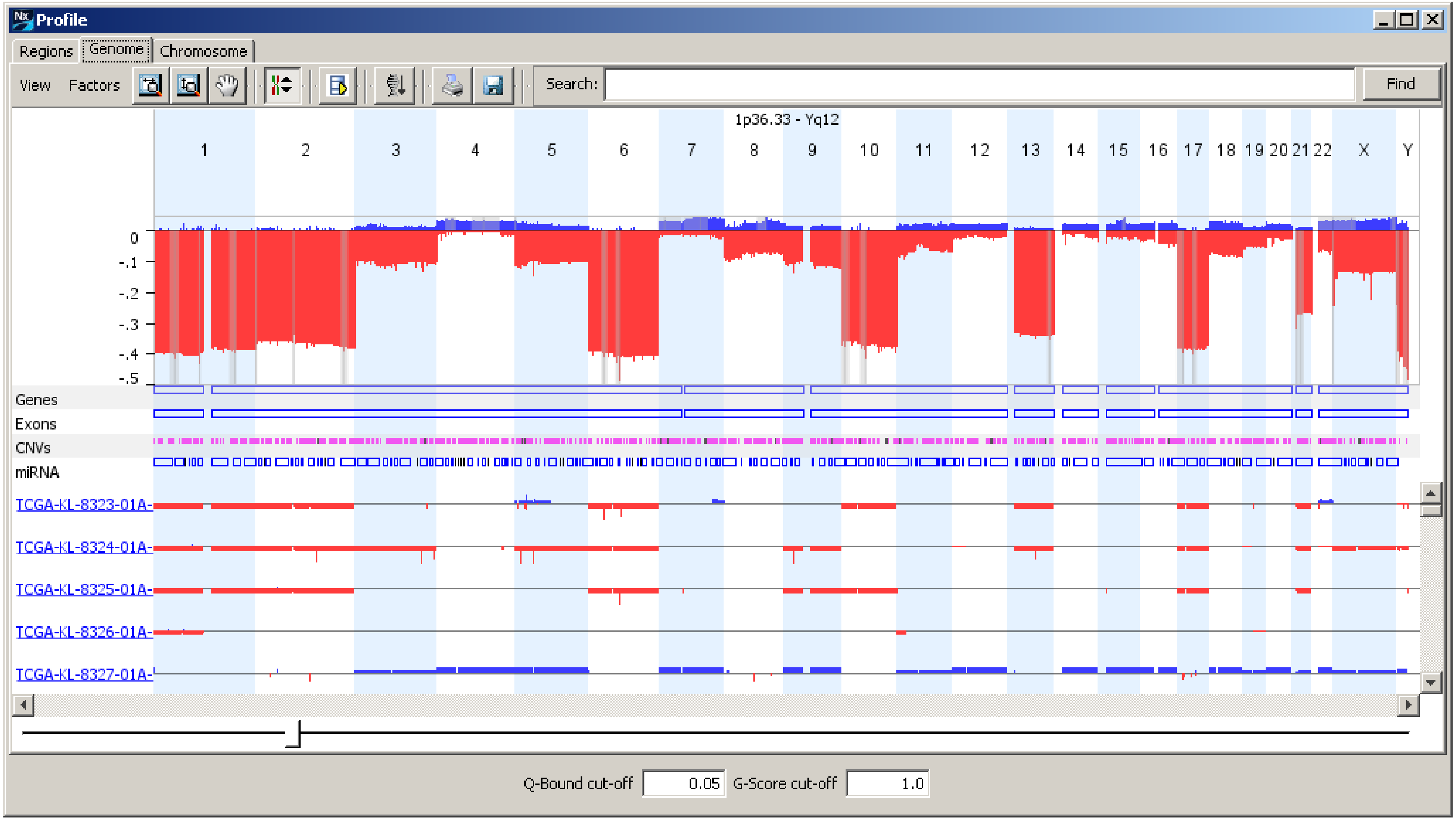
GISTIC Results



Region	Extended Region	Type	Q-Bound	G-Score	% of CNV Overlap
chr4:37,874,548-37,901,732	chr4:22,370,696-49,309,867	CN Gain	0.02	2.433	100
chr4:114,290,717-114,317,844	chr4:86,982,431-155,526,202	CN Gain	0.046	2.148	100
chr5:1,114,344-1,224,819	chr5:1,045,016-1,602,048	CN Gain	0.039	2.217	100
chr7:101,100,758-101,147,413	chr7:61,829,069-121,899,445	CN Gain	0.019	2.894	100
chr7:13,986,863-13,991,819	chr7:0-57,950,944	CN Gain	0.039	2.193	100
chr8:101,329,453-103,273,593	chr8:82,537,767-105,512,220	CN Gain	0.019	2.58	58.224
chr15:64,050,707-64,245,807	chr15:59,698,546-68,148,852	CN Gain	0.019	2.567	37.775
chr22:51,029,323-51,036,784	chr22:17,241,078-51,304,566	CN Gain	0.039	2.202	100
chrX:147,518,948-147,564,009	chrX:143,271,422-154,929,403	CN Gain	0.019	2.727	20.195
chrX:45,965,212-46,032,977	chrX:2,684,878-58,317,029	CN Gain	0.034	2.263	100
chr1:111,378,980-111,379,483	chr1:110,245,562-112,693,006	CN Loss	2.01E-13	28.208	100
chr1:196,823,770-196,907,402	chr1:185,055,969-202,735,388	CN Loss	2.01E-13	25.989	100
chr1:51,086,901-51,448,283	chr1:40,006,946-62,523,627	CN Loss	0.037	15.119	30.593
chr2:213,186,444-213,190,071	chr2:208,359,406-226,191,300	CN Loss	2.01E-13	25.52	100
chr2:0-113,423	chr2:0-3,769,089	CN Loss	2.01E-13	25.193	91.183
chr2:91,794,849-91,844,183	chr2:91,704,724-92,193,296	CN Loss	8.70E-10	22.144	100
chr6:74,592,335-74,599,375	chr6:67,052,760-77,436,346	CN Loss	2.01E-13	31.788	100
chr6:36,599,687-36,688,808	chr6:31,453,655-46,898,826	CN Loss	2.01E-13	26.913	67.489
chr10:58,521,579-58,526,368	chr10:47,067,454-61,358,062	CN Loss	2.01E-13	26.942	100
chr10:6,663,413-6,664,229	chr10:0-20,274,925	CN Loss	2.13E-13	24.704	100
chr13:104,276,598-104,279,...	chr13:99,694,090-109,554,181	CN Loss	2.47E-11	23.157	100
chr17:39,519,402-39,524,402	chr17:39,433,067-49,903,094	CN Loss	2.01E-13	26.124	100
chr17:15,038,012-15,056,753	chr17:0-16,850,302	CN Loss	2.01E-13	25.791	100
chr21:10,988,131-10,988,413	chr21:9,764,385-11,018,120	CN Loss	9.09E-08	20.708	100

Q-Bound cut-off 0.05 G-Score cut-off 1.0

GISTIC – Genome View


Profile
Regions
Genome
Chromosome
View
Factors
Search:
Find
1p36.33 - Yq12
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CNVs
miRNA
TCGA-KL-8323-01A-
TCGA-KL-8324-01A-
TCGA-KL-8325-01A-
TCGA-KL-8326-01A-
TCGA-KL-8327-01A-

Q-Bound cut-off 0.05 G-Score cut-off 1.0

Which algorithm should I use?

GISTIC results may be more sensitive at capturing lower frequency significant regions

STAC will give more correct results for sex chromosomes and manually altered regions, if applicable

Overall, the two methods will often yield similar/overlapping results; a combined approach may be most thorough.

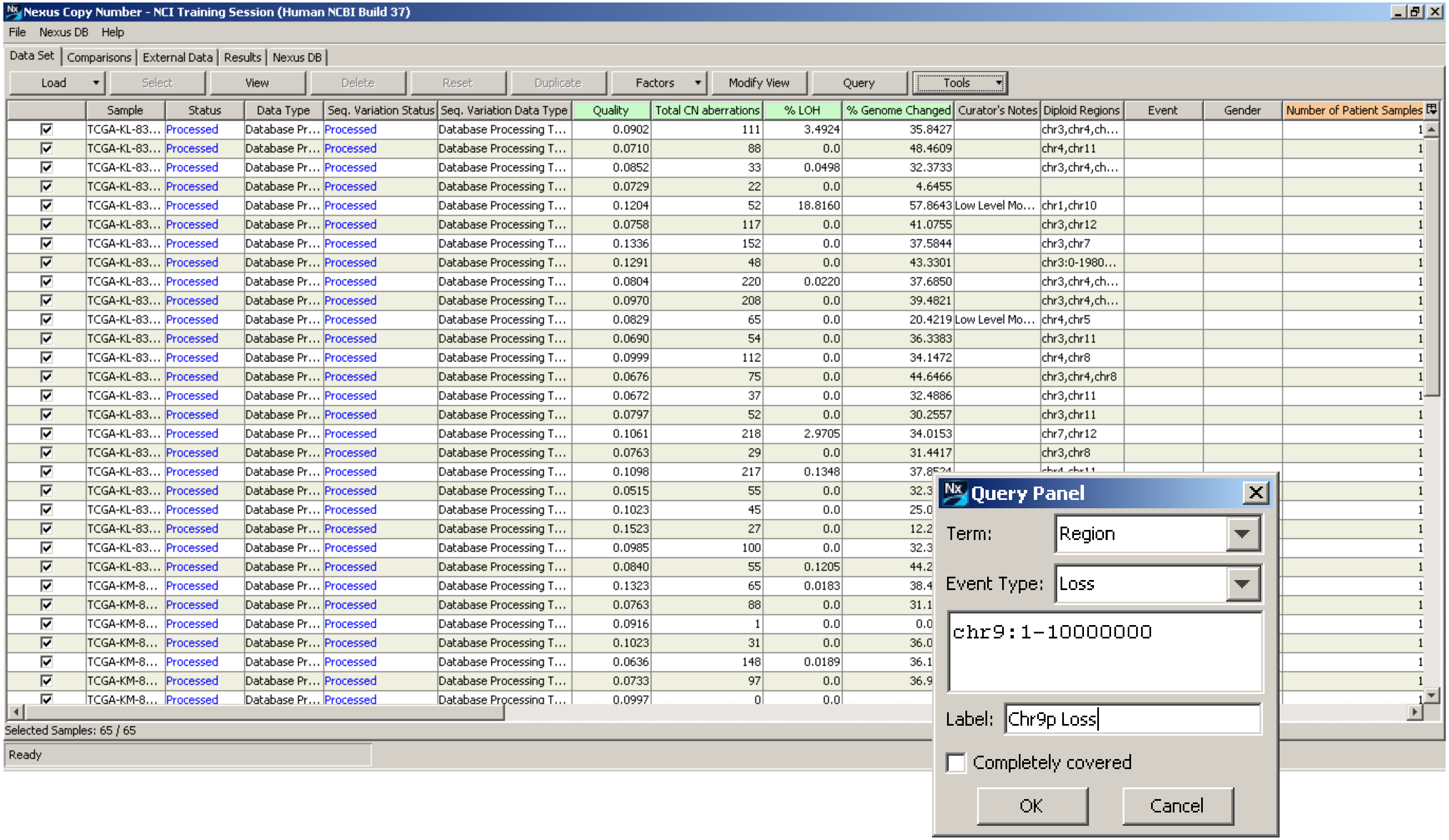
Identifying significant co-occurring alterations

Which changes significantly occur with a known change of interest

Concordance – Data Set Tab

- Search for one or more genes, regions, etc. within the project
- Returns the status of each sample for each query term as well as aggregate summary
- Select samples of interest for result export and data analysis
- Provides Frequency of each event for each of the queried terms

Concordance – Chr9p Loss



	Sample	Status	Data Type	Seq. Variation Status	Seq. Variation Data Type	Quality	Total CN aberrations	% LOH	% Genome Changed	Curator's Notes	Diploid Regions	Event	Gender	Number of Patient Samples
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0902	111	3.4924	35.8427		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0710	88	0.0	48.4609		chr4,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0852	33	0.0498	32.3733		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0729	22	0.0	4.6455					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1204	52	18.8160	57.8643	Low Level Mo…	chr1,chr10			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0758	117	0.0	41.0755		chr3,chr12			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1336	152	0.0	37.5844		chr3,chr7			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1291	48	0.0	43.3301		chr3:0-1980…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0804	220	0.0220	37.6850		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0970	208	0.0	39.4821		chr3,chr4,ch…			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0829	65	0.0	20.4219	Low Level Mo…	chr4,chr5			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0690	54	0.0	36.3383		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0999	112	0.0	34.1472		chr4,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0676	75	0.0	44.6466		chr3,chr4,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0672	37	0.0	32.4886		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0797	52	0.0	30.2557		chr3,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1061	218	2.9705	34.0153		chr7,chr12			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0763	29	0.0	31.4417		chr3,chr8			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1098	217	0.1348	37.85		chr4,chr11			1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0515	55	0.0	32.3					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1023	45	0.0	25.0					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.1523	27	0.0	12.2					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0985	100	0.0	32.3					1
☑	TCGA-KL-83…	Processed	Database Pr…	Processed	Database Processing T…	0.0840	55	0.1205	44.2					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.1323	65	0.0183	38.4					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0763	88	0.0	31.1					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0916	1	0.0	0.0					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.1023	31	0.0	36.0					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0636	148	0.0189	36.1					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0733	97	0.0	36.9					1
☑	TCGA-KM-8…	Processed	Database Pr…	Processed	Database Processing T…	0.0997	0	0.0						1

Selected Samples: 65 / 65

Ready

Term: Region

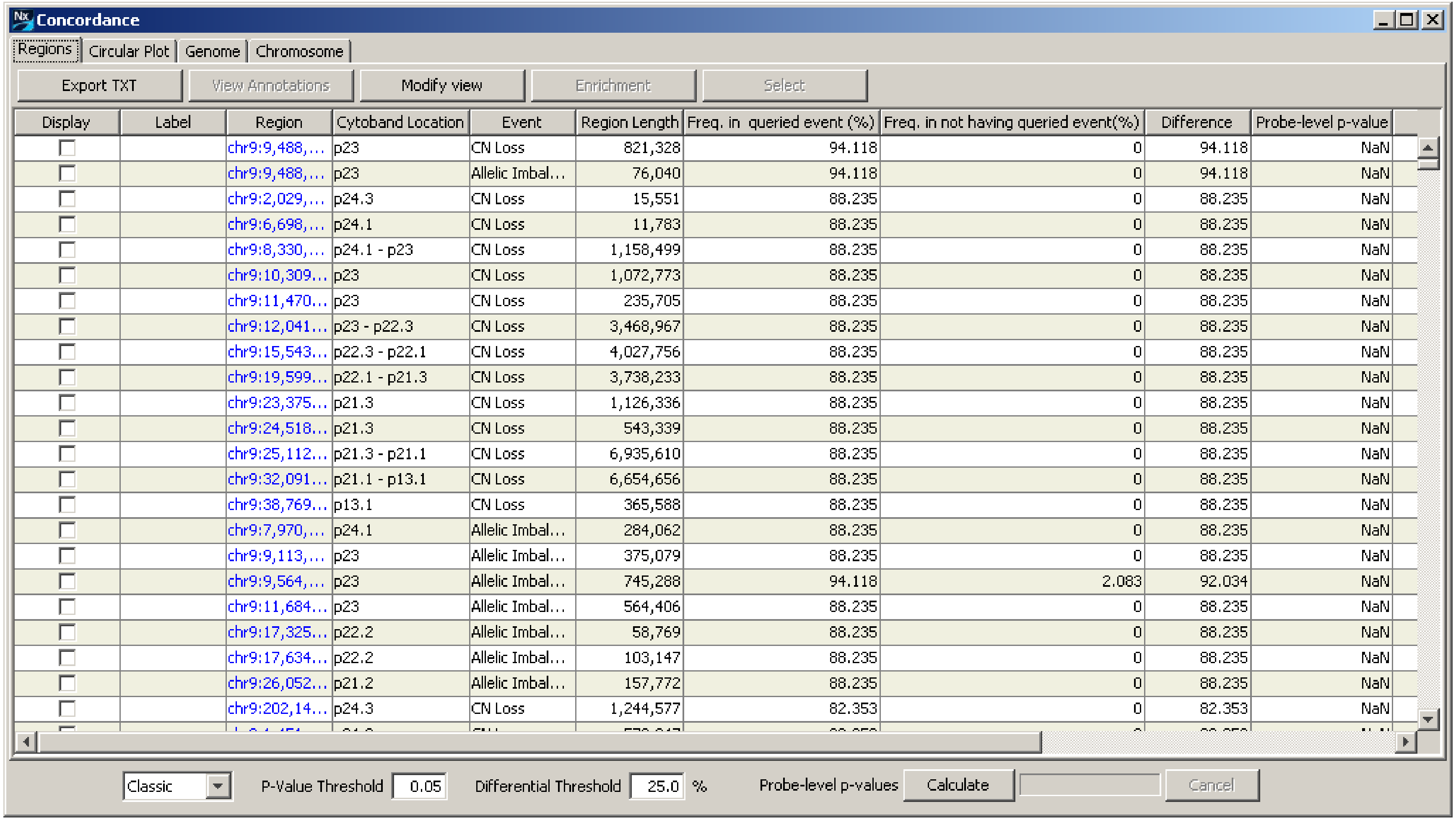
Event Type: Loss

chr9:1-10000000

Label: Chr9p Loss

☐ Completely covered

Chr9p Concordance - Regions



Display	Label	Region	Cytoband Location	Event	Region Length	Freq. in queried event (%)	Freq. in not having queried event(%)	Difference	Probe-level p-value
☐		chr9:9,488,...	p23	CN Loss	821,328	94.118	0	94.118	NaN
☐		chr9:9,488,...	p23	Allelic Imbal...	76,040	94.118	0	94.118	NaN
☐		chr9:2,029,...	p24.3	CN Loss	15,551	88.235	0	88.235	NaN
☐		chr9:6,698,...	p24.1	CN Loss	11,783	88.235	0	88.235	NaN
☐		chr9:8,330,...	p24.1 - p23	CN Loss	1,158,499	88.235	0	88.235	NaN
☐		chr9:10,309...	p23	CN Loss	1,072,773	88.235	0	88.235	NaN
☐		chr9:11,470...	p23	CN Loss	235,705	88.235	0	88.235	NaN
☐		chr9:12,041...	p23 - p22.3	CN Loss	3,468,967	88.235	0	88.235	NaN
☐		chr9:15,543...	p22.3 - p22.1	CN Loss	4,027,756	88.235	0	88.235	NaN
☐		chr9:19,599...	p22.1 - p21.3	CN Loss	3,738,233	88.235	0	88.235	NaN
☐		chr9:23,375...	p21.3	CN Loss	1,126,336	88.235	0	88.235	NaN
☐		chr9:24,518...	p21.3	CN Loss	543,339	88.235	0	88.235	NaN
☐		chr9:25,112...	p21.3 - p21.1	CN Loss	6,935,610	88.235	0	88.235	NaN
☐		chr9:32,091...	p21.1 - p13.1	CN Loss	6,654,656	88.235	0	88.235	NaN
☐		chr9:38,769...	p13.1	CN Loss	365,588	88.235	0	88.235	NaN
☐		chr9:7,970,...	p24.1	Allelic Imbal...	284,062	88.235	0	88.235	NaN
☐		chr9:9,113,...	p23	Allelic Imbal...	375,079	88.235	0	88.235	NaN
☐		chr9:9,564,...	p23	Allelic Imbal...	745,288	94.118	2.083	92.034	NaN
☐		chr9:11,684...	p23	Allelic Imbal...	564,406	88.235	0	88.235	NaN
☐		chr9:17,325...	p22.2	Allelic Imbal...	58,769	88.235	0	88.235	NaN
☐		chr9:17,634...	p22.2	Allelic Imbal...	103,147	88.235	0	88.235	NaN
☐		chr9:26,052...	p21.2	Allelic Imbal...	157,772	88.235	0	88.235	NaN
☐		chr9:202,14...	p24.3	CN Loss	1,244,577	82.353	0	82.353	NaN

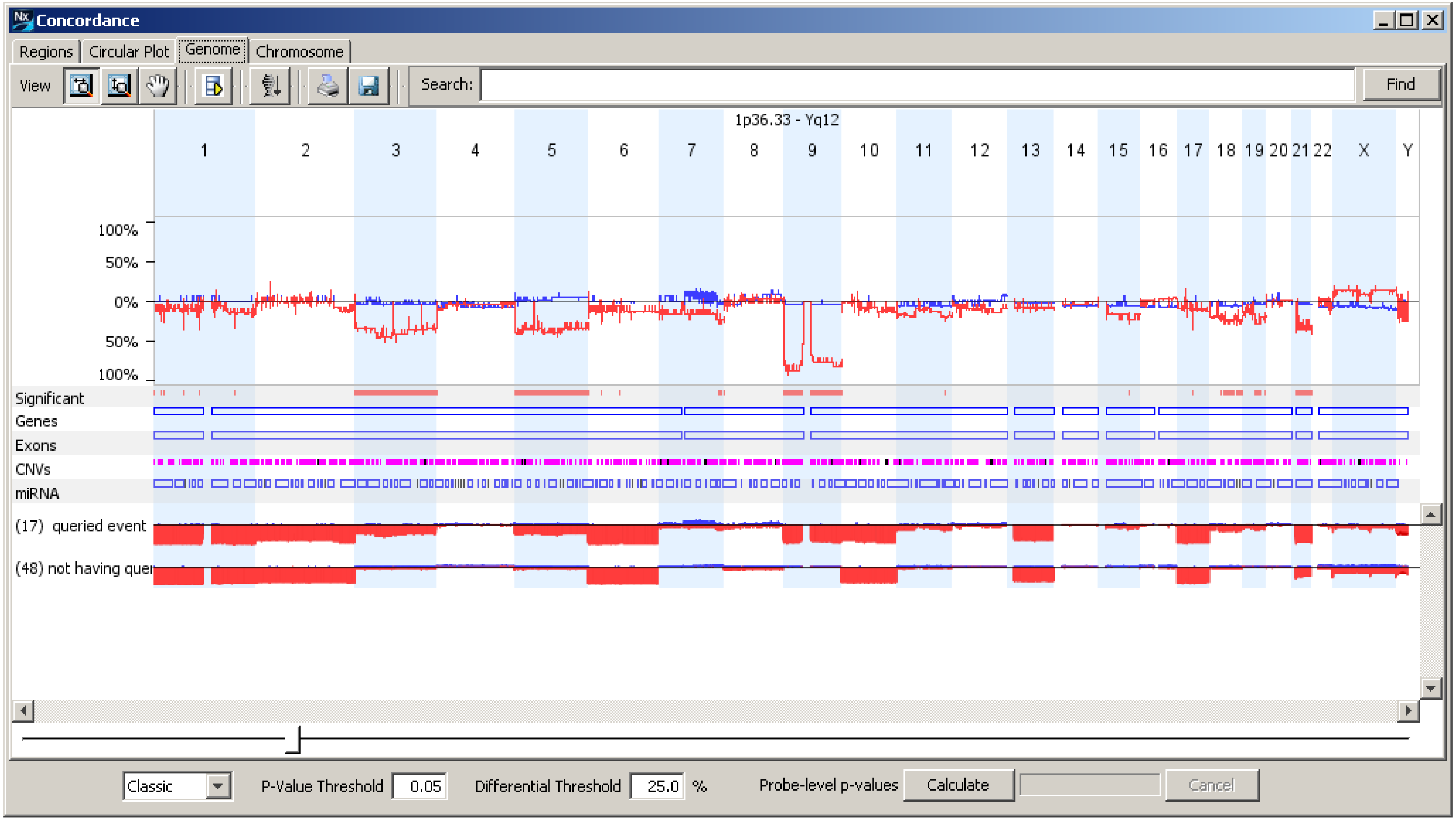
Classic

P-Value Threshold 0.05

Differential Threshold 25.0 %

Probe-level p-values

Chr9p Concordance - Genome



Concordance
Regions
Circular Plot
Genome
Chromosome
View
Search:
Find
1p36.33 - Yq12
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
100%
50%
0%
50%
100%
Significant
Genes
Exons
CNVs
miRNA
(17) queried event
(48) not having quer
Calculate
Cancel

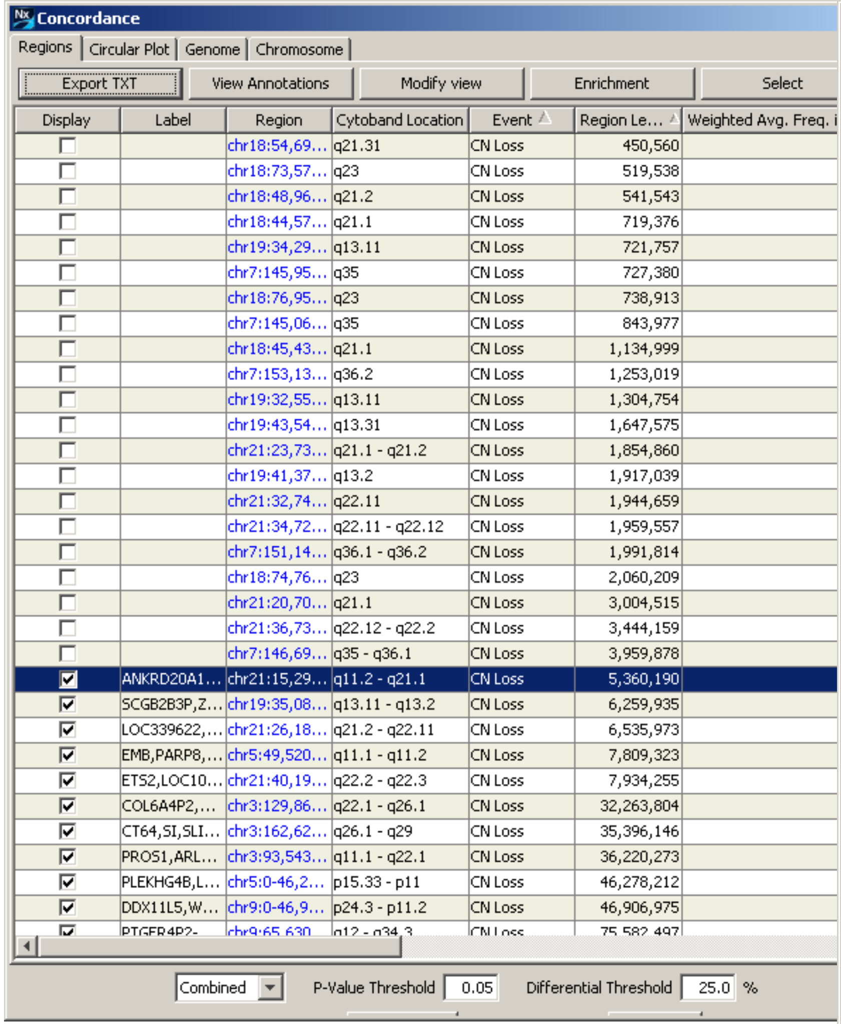
Classic

P-Value Threshold 0.05

Differential Threshold 25.0 %

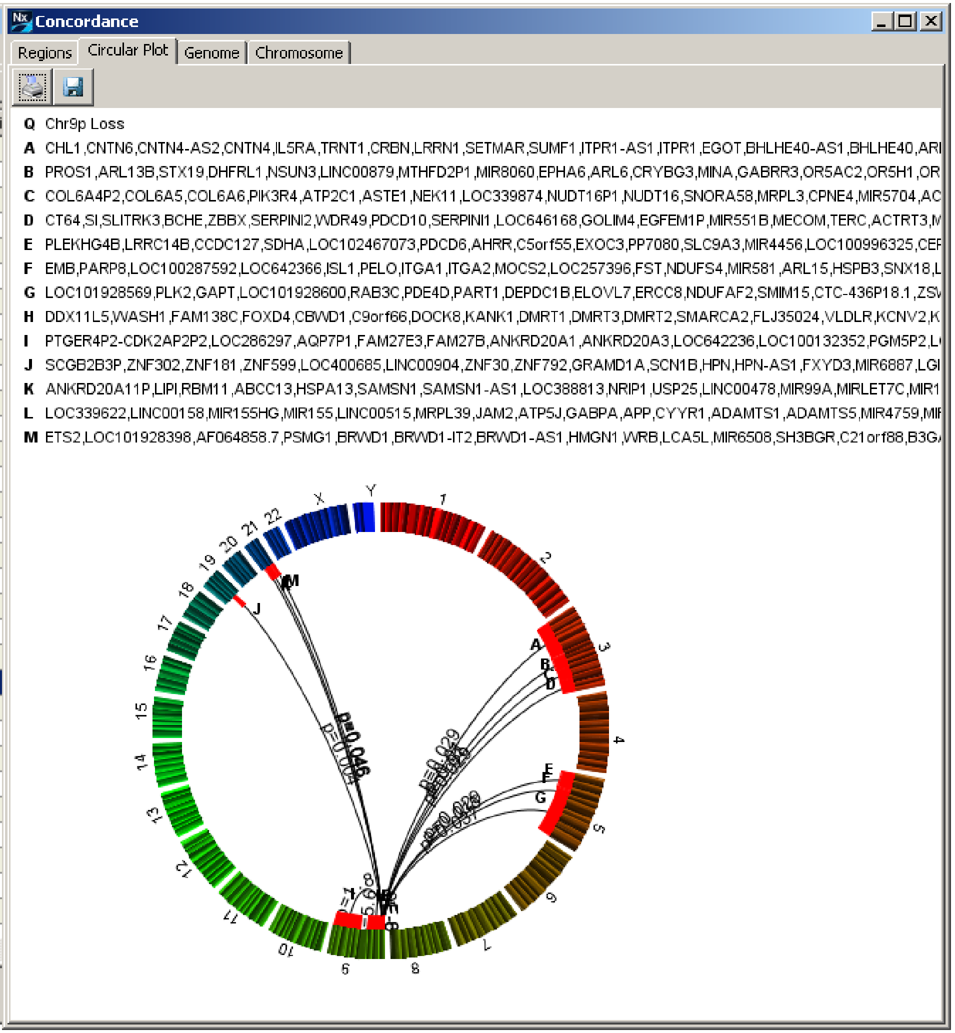
Probe-level p-values

Chr9p Concordance – Circular Plot



Display	Label	Region	Cytoband Location	Event	Region Le...	Weighted Avg. Freq. i
☐		chr18:54,69...	q21.31	CN Loss	450,560	
☐		chr18:73,57...	q23	CN Loss	519,538	
☐		chr18:48,96...	q21.2	CN Loss	541,543	
☐		chr18:44,57...	q21.1	CN Loss	719,376	
☐		chr19:34,29...	q13.11	CN Loss	721,757	
☐		chr7:145,95...	q35	CN Loss	727,380	
☐		chr18:76,95...	q23	CN Loss	738,913	
☐		chr7:145,06...	q35	CN Loss	843,977	
☐		chr18:45,43...	q21.1	CN Loss	1,134,999	
☐		chr7:153,13...	q36.2	CN Loss	1,253,019	
☐		chr19:32,55...	q13.11	CN Loss	1,304,754	
☐		chr19:43,54...	q13.31	CN Loss	1,647,575	
☐		chr21:23,73...	q21.1 - q21.2	CN Loss	1,854,860	
☐		chr19:41,37...	q13.2	CN Loss	1,917,039	
☐		chr21:32,74...	q22.11	CN Loss	1,944,659	
☐		chr21:34,72...	q22.11 - q22.12	CN Loss	1,959,557	
☐		chr7:151,14...	q36.1 - q36.2	CN Loss	1,991,814	
☐		chr18:74,76...	q23	CN Loss	2,060,209	
☐		chr21:20,70...	q21.1	CN Loss	3,004,515	
☐		chr21:36,73...	q22.12 - q22.2	CN Loss	3,444,159	
☐		chr7:146,69...	q35 - q36.1	CN Loss	3,959,878	
☑	ANKRD20A1...	chr21:15,29...	q11.2 - q21.1	CN Loss	5,360,190	
☑	SCGB2B3P,Z...	chr19:35,08...	q13.11 - q13.2	CN Loss	6,259,935	
☑	LOC339622,...	chr21:26,18...	q21.2 - q22.11	CN Loss	6,535,973	
☑	EMB,PARP8,...	chr5:49,520...	q11.1 - q11.2	CN Loss	7,809,323	
☑	ETS2,LOC10...	chr21:40,19...	q22.2 - q22.3	CN Loss	7,934,255	
☑	COL6A4P2,...	chr3:129,86...	q22.1 - q26.1	CN Loss	32,263,804	
☑	CT64,SI,SLI...	chr3:162,62...	q26.1 - q29	CN Loss	35,396,146	
☑	PROS1,ARL...	chr3:93,543...	q11.1 - q22.1	CN Loss	36,220,273	
☑	PLEKHG4B,L...	chr5:0-46,2...	p15.33 - p11	CN Loss	46,278,212	
☑	DDX11L5,W...	chr9:0-46,9...	p24.3 - p11.2	CN Loss	46,906,975	
☑	PTGER4P2-	chr9:65,630	q12 - q34.3	CN Loss	75,582,497	

Combined ▾ P-Value Threshold 0.05 Differential Threshold 25.0 %

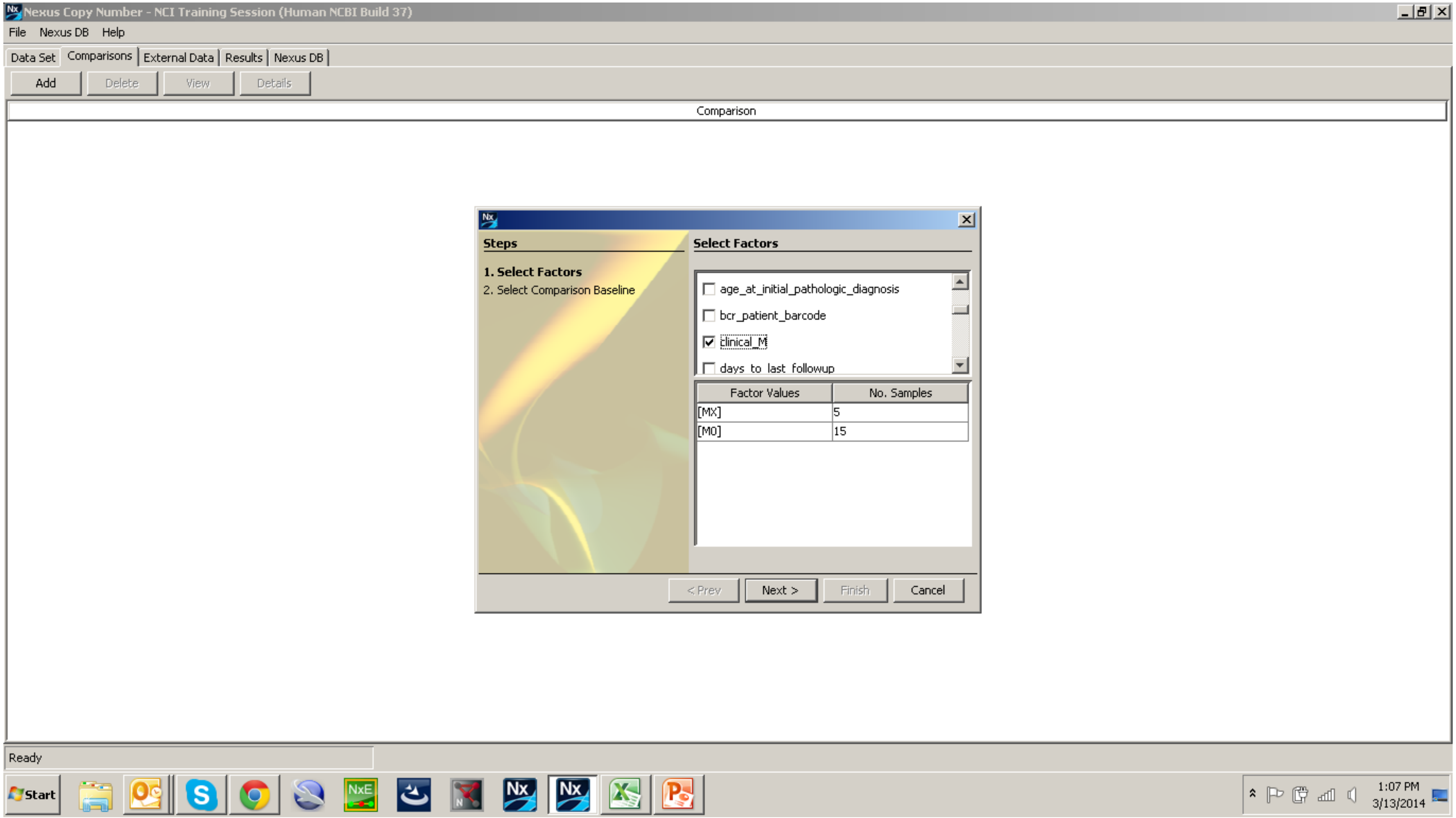


Q Chr9p Loss
A CHL1,CNTN6,CNTN4-AS2,CNTN4,IL5RA,TRNT1,CRBN,LRRN1,SETMAR,SUMF1,ITPR1-AS1,ITPR1,EGOT,BHLHE40-AS1,BHLHE40,ARI
B PROS1,ARL13B,STX19,DHFRL1,NSUN3,LINC00879,MTHFD2P1,MIR8060,EPHA6,ARL6,CRYBG3,MINA,GABRR3,OR5AC2,OR5H1,OR
C COL6A4P2,COL6A5,COL6A6,PIK3R4,ATP2C1,ASTE1,NEK11,LOC339874,NUDT16P1,NUDT16,SNORA58,MRPL3,CPNE4,MIR5704,AC
D CT64,SI,SLITRK3,BCHE,ZBBX,SERPINI2,WDR49,PDCD10,SERPINI1,LOC646168,GOLIM4,EGFEM1P,MIR551B,MECOM,TERC,ACTRT3,M
E PLEKHG4B,LRRC14B,CCDC127,SDHA,LOC102467073,PDCD6,AHRR,C5orf55,EXOC3,PP7080,SLC9A3,MIR4456,LOC100996325,CEF
F EMB,PARP8,LOC100287592,LOC642366,ISL1,PELO,ITGA1,ITGA2,MOCS2,LOC257396,FST,NDUFS4,MIR581,ARL15,HSPB3,SNX18,L
G LOC101928569,PLK2,GAPT,LOC101928600,RAB3C,PDE4D,PART1,DEPDC1B,ELOVL7,ERCC8,NDUFAF2,SMIM15,CTC-436P18.1,ZSW
H DDX11L5,WASH1,FAM138C,FOXD4,CBWD1,C9orf66,DOCK8,KANK1,DMRT1,DMRT3,DMRT2,SMARCA2,FLJ35024,VLDLR,KCNV2,K
I PTGER4P2-CDK2AP2P2,LOC286297,AQP7P1,FAM27E3,FAM27B,ANKRD20A1,ANKRD20A3,LOC642236,LOC100132352,PGM5P2,L
J SCGB2B3P,ZNF302,ZNF181,ZNF599,LOC400685,LINC00904,ZNF30,ZNF792,GRAMD1A,SCN1B,HPN,HPN-AS1,FXYD3,MIR6887,LGI
K ANKRD20A11P,LIPI,RBM11,ABCC13,HSPA13,SAMSN1,SAMSN1-AS1,LOC388813,NRIP1,USP25,LINC00478,MIR99A,MIRLET7C,MIR1
L LOC339622,LINC00158,MIR155HG,MIR155,LINC00515,MRPL39,JAM2,ATP5J,GABPA,APP,CYYR1,ADAMTS1,ADAMTS5,MIR4759,MIF
M ETS2,LOC101928398,AF064858.7,PSMG1,BRWD1,BRWD1-IT2,BRWD1-AS1,HMGN1,WRB,LCA5L,MIR6508,SH3BGR,C21orf88,B3G

Comparing groups to find significant differences

What changes are significant and unique to each group?

Comparison – Metastasis (Clinical_M)

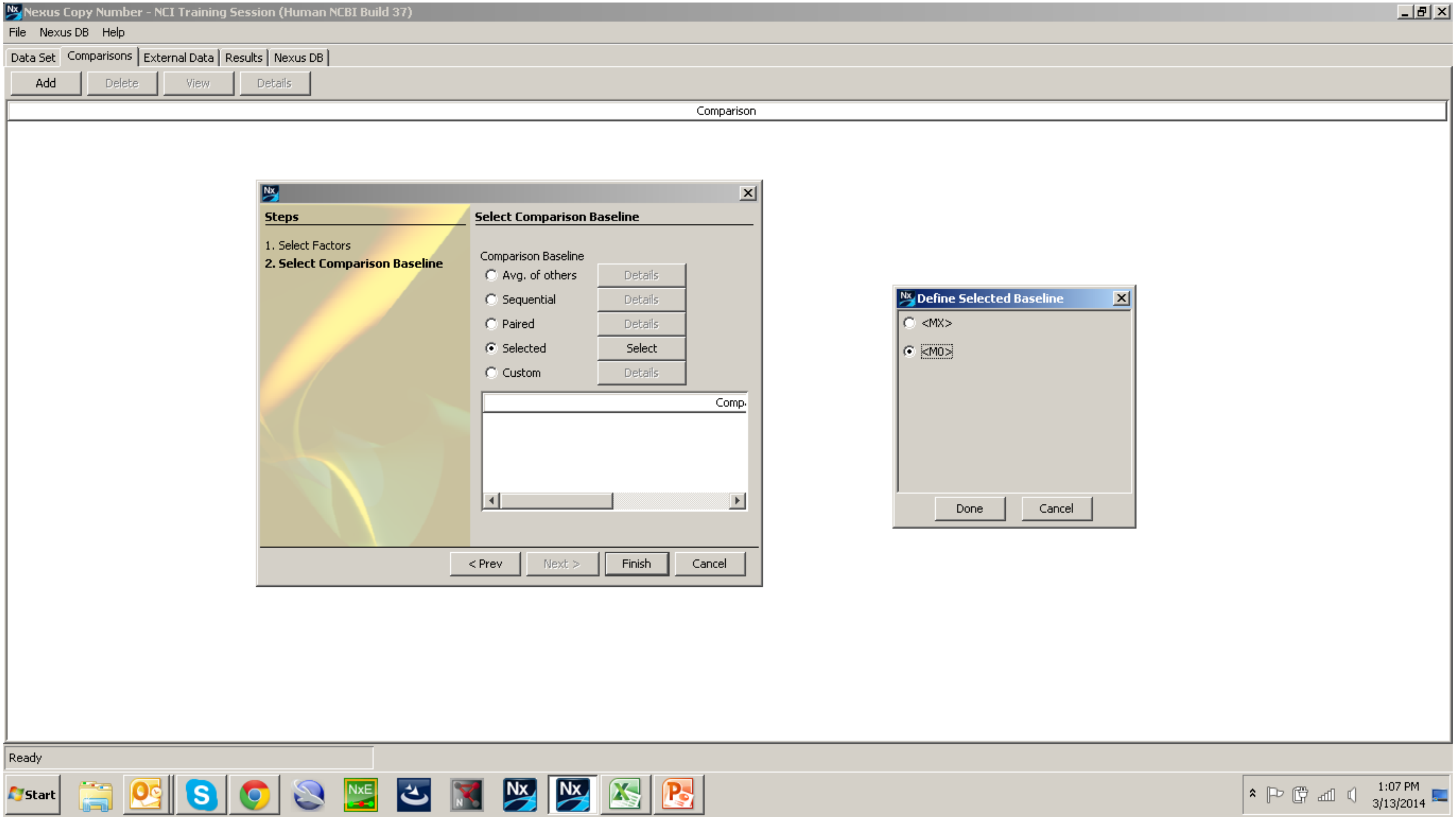


Nexus Copy Number - NCI Training Session (Human NCBI Build 37)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Results
Nexus DB
Add
Delete
View
Details
Comparison
Steps
1. Select Factors
2. Select Comparison Baseline
Select Factors
< Prev
Next >
Finish
Cancel
Ready
Start
1:07 PM
3/13/2014

☐ age_at_initial_pathologic_diagnosis
☐ bcr_patient_barcode
☑ clinical_M
☐ days_to_last_followup

Factor Values	No. Samples
[MX]	5
[M0]	15

Comparison – M0 v MX



Nexus Copy Number - NCI Training Session (Human NCBI Build 37)

Steps

1. Select Factors
2. **Select Comparison Baseline**

Select Comparison Baseline

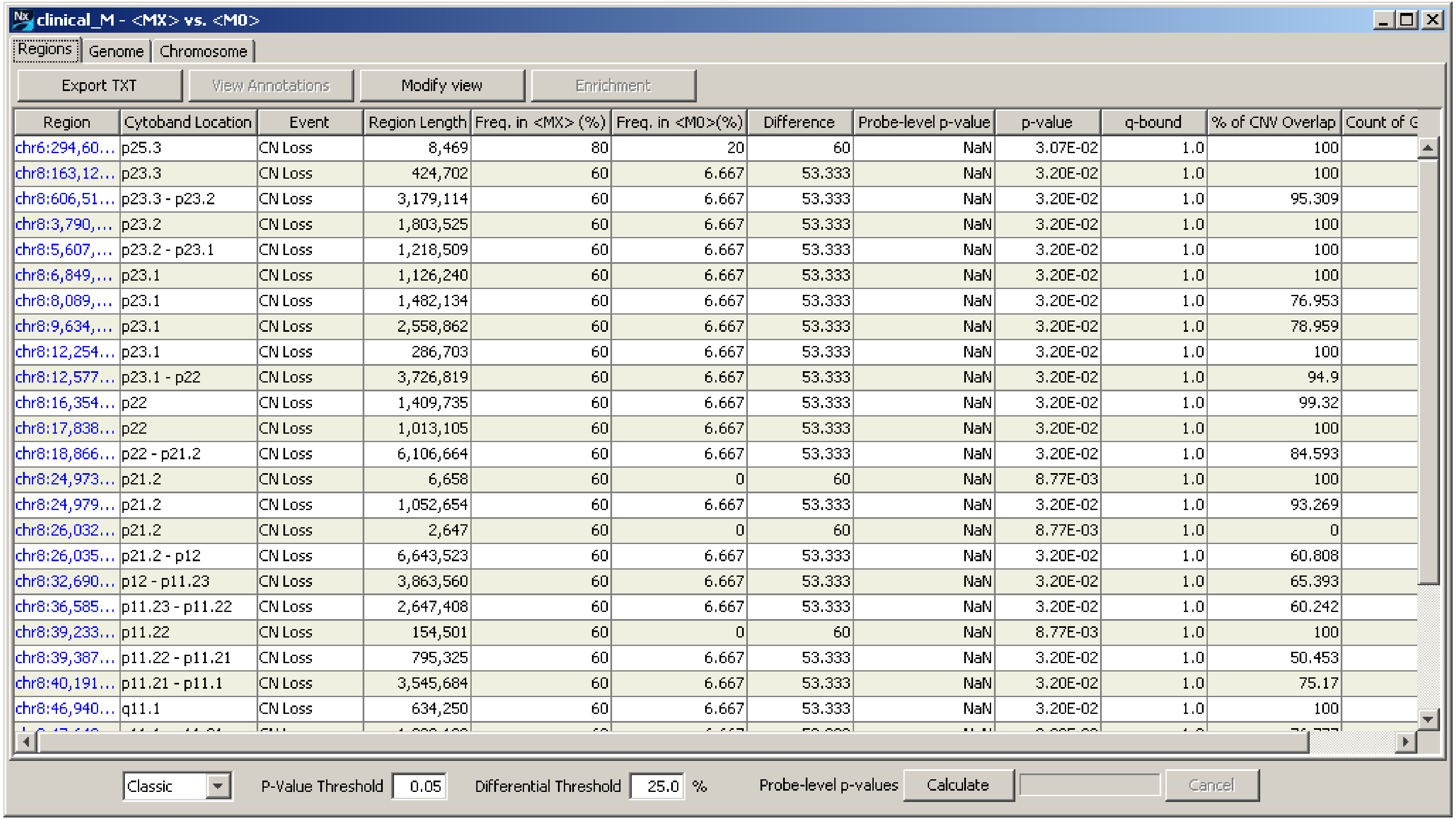
Comparison Baseline
- ○ Avg. of others
- ○ Sequential
- ○ Paired
- ● Selected
- ○ Custom

Define Selected Baseline
- ○ <MX>
- ● <M0>

Ready

File
Nexus DB
Help
Data Set
Comparisons
External Data
Results
Nexus DB
Add
Delete
View
Details
Comparison
Details
Select
Comp.
< Prev
Next >
Finish
Cancel
Done
Cancel
Start
1:07 PM
3/13/2014

Comparison – M0 v MX - Regions



Region	Cytoband Location	Event	Region Length	Freq. in <MX> (%)	Freq. in <M0>(%)	Difference	Probe-level p-value	p-value	q-bound	% of CNV Overlap	Count of G
chr6:294,60...	p25.3	CN Loss	8,469	80	20	60	NaN	3.07E-02	1.0	100	
chr8:163,12...	p23.3	CN Loss	424,702	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:606,51...	p23.3 - p23.2	CN Loss	3,179,114	60	6.667	53.333	NaN	3.20E-02	1.0	95.309	
chr8:3,790,...	p23.2	CN Loss	1,803,525	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:5,607,...	p23.2 - p23.1	CN Loss	1,218,509	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:6,849,...	p23.1	CN Loss	1,126,240	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:8,089,...	p23.1	CN Loss	1,482,134	60	6.667	53.333	NaN	3.20E-02	1.0	76.953	
chr8:9,634,...	p23.1	CN Loss	2,558,862	60	6.667	53.333	NaN	3.20E-02	1.0	78.959	
chr8:12,254...	p23.1	CN Loss	286,703	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:12,577...	p23.1 - p22	CN Loss	3,726,819	60	6.667	53.333	NaN	3.20E-02	1.0	94.9	
chr8:16,354...	p22	CN Loss	1,409,735	60	6.667	53.333	NaN	3.20E-02	1.0	99.32	
chr8:17,838...	p22	CN Loss	1,013,105	60	6.667	53.333	NaN	3.20E-02	1.0	100	
chr8:18,866...	p22 - p21.2	CN Loss	6,106,664	60	6.667	53.333	NaN	3.20E-02	1.0	84.593	
chr8:24,973...	p21.2	CN Loss	6,658	60	0	60	NaN	8.77E-03	1.0	100	
chr8:24,979...	p21.2	CN Loss	1,052,654	60	6.667	53.333	NaN	3.20E-02	1.0	93.269	
chr8:26,032...	p21.2	CN Loss	2,647	60	0	60	NaN	8.77E-03	1.0	0	
chr8:26,035...	p21.2 - p12	CN Loss	6,643,523	60	6.667	53.333	NaN	3.20E-02	1.0	60.808	
chr8:32,690...	p12 - p11.23	CN Loss	3,863,560	60	6.667	53.333	NaN	3.20E-02	1.0	65.393	
chr8:36,585...	p11.23 - p11.22	CN Loss	2,647,408	60	6.667	53.333	NaN	3.20E-02	1.0	60.242	
chr8:39,233...	p11.22	CN Loss	154,501	60	0	60	NaN	8.77E-03	1.0	100	
chr8:39,387...	p11.22 - p11.21	CN Loss	795,325	60	6.667	53.333	NaN	3.20E-02	1.0	50.453	
chr8:40,191...	p11.21 - p11.1	CN Loss	3,545,684	60	6.667	53.333	NaN	3.20E-02	1.0	75.17	
chr8:46,940...	q11.1	CN Loss	634,250	60	6.667	53.333	NaN	3.20E-02	1.0	100	

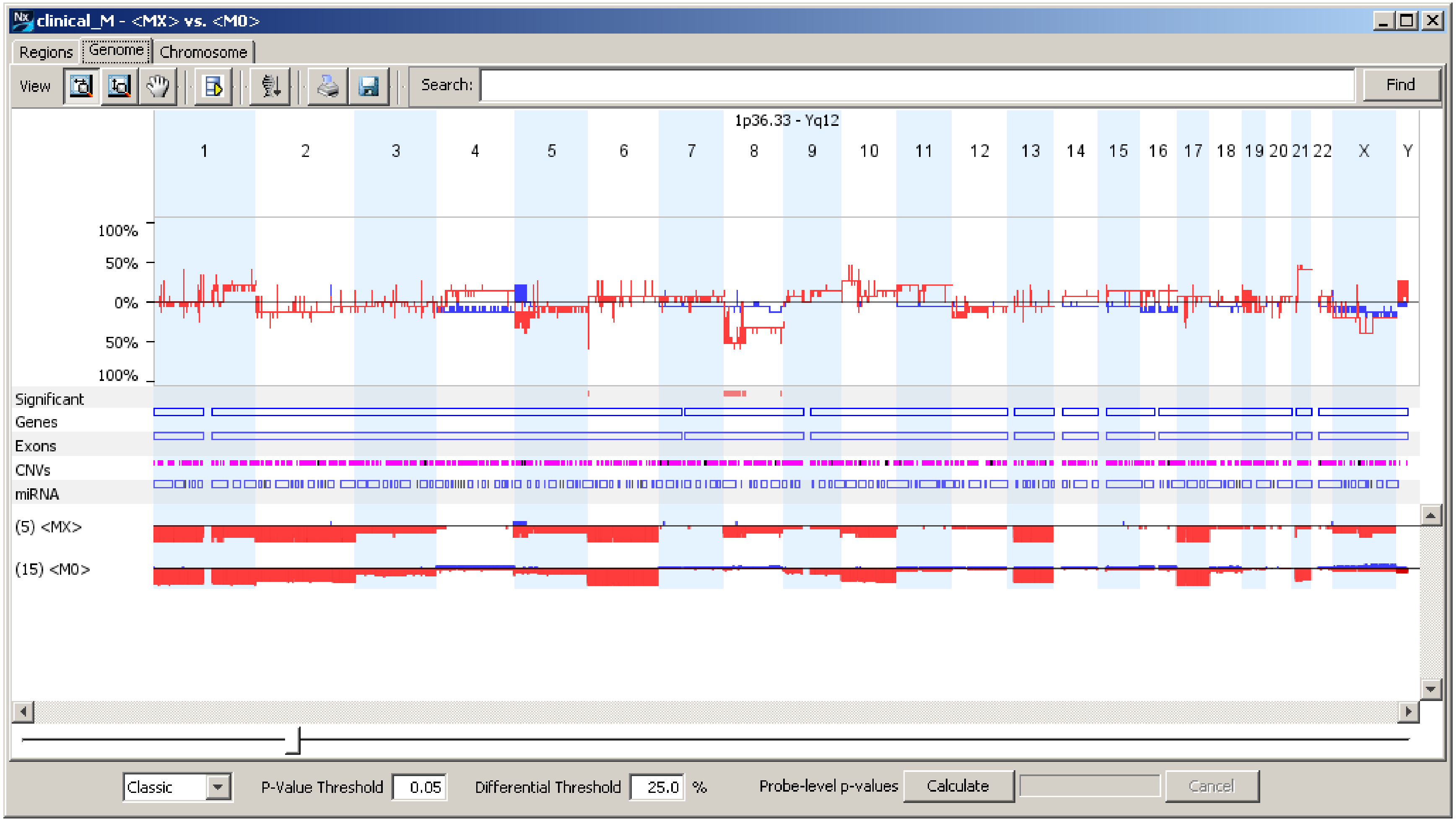
Classic

P-Value Threshold 0.05

Differential Threshold 25.0 %

Probe-level p-values

Comparison – M0 v MX - Genome


clinical_M - <MX> vs. <M0>
Regions
Genome
Chromosome
View
Search:
Find
1p36.33 - Yq12
1
2
3
4
5
6
7
8
9
10
11
12
13
14
15
16
17
18
19
20
21
22
X
Y
100%
50%
0%
50%
100%
Significant
Genes
Exons
CNVs
miRNA
(5) <MX>
(15) <M0>
Calculate
Cancel

Classic

P-Value Threshold 0.05

Differential Threshold 25.0 %

Probe-level p-values

Enrichment Analysis

Gene enrichment analysis on the whole genome or selected regions of interest

Which pathways or processes are enriched in my data set?

Enrichment on the whole genome

- For germline studies or samples with few changes

Enrichment on selected regions of interest

- Aggregate (Frequency)
- Significant Peaks (STAC)
- GISTIC
- Comparisons
- Concordance

Enrichment on Selected Regions

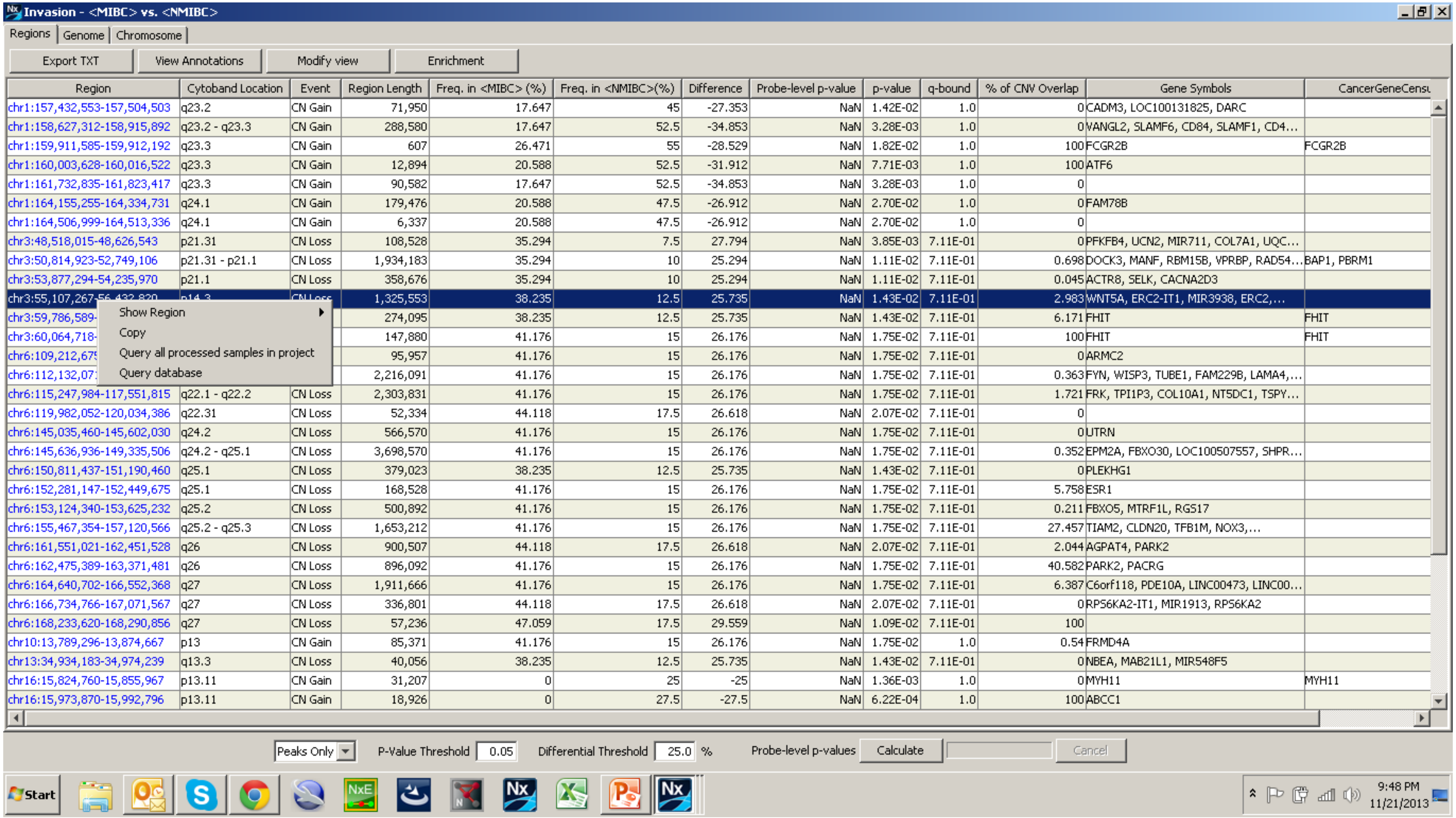
Identifies Gene Ontology (GO) terms enriched with copy number change in the user defined regions

  - Predefined highlighted area(s) of interest

- Displays significantly overrepresented GO terms
  - P-Value displays standard p-value: probability of each particular gene being present in the set treated independently of others
  - MP P-Value displays Markov process p-value: probability of each gene being present in the set takes into account genomic location

Querying Results in Nexus DB

Has this change been observed before in another data set?



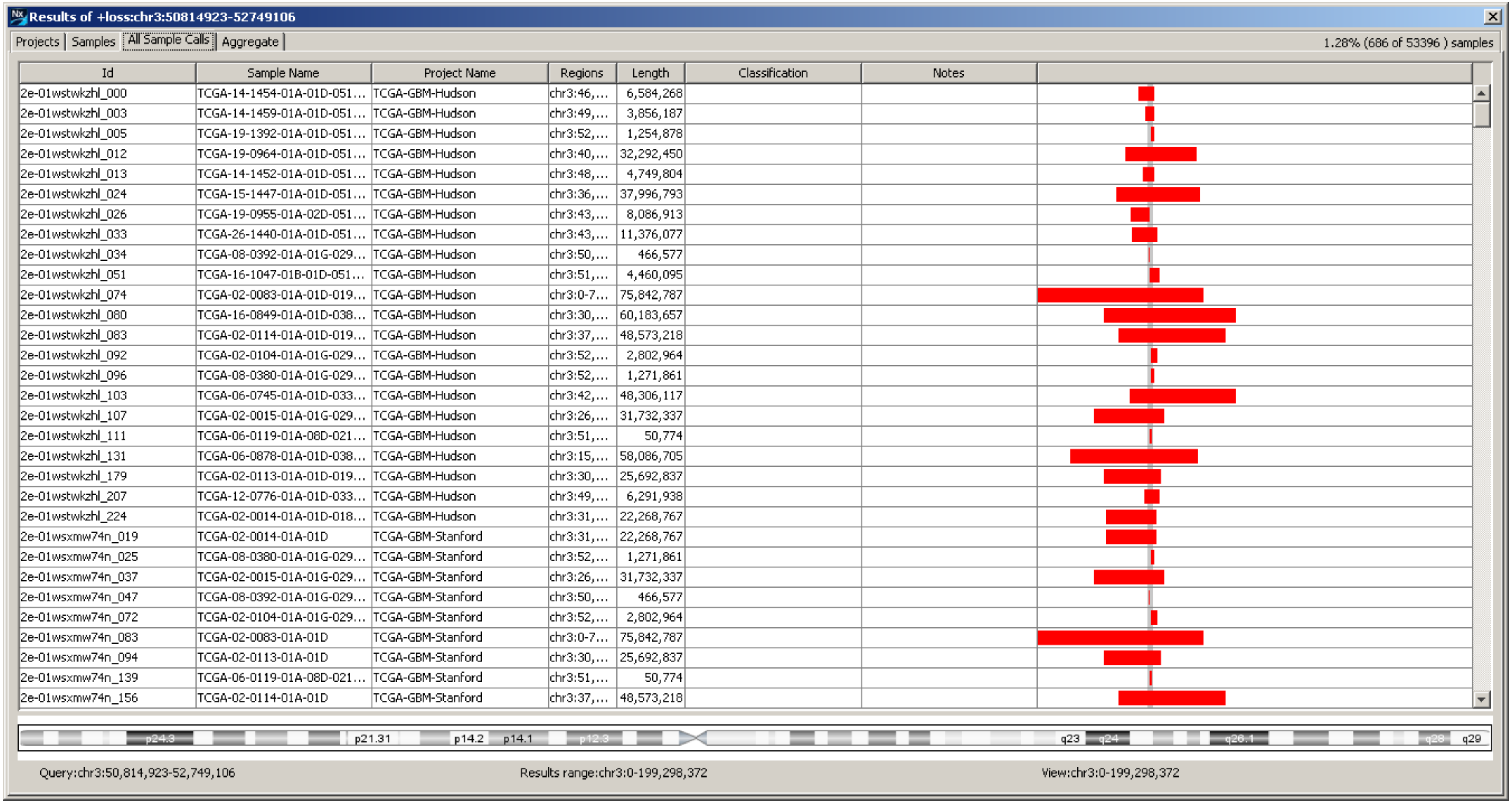
Region	Cytoband Location	Event	Region Length	Freq. in <MIBC> (%)	Freq. in <NMIBC>(%)	Difference	Probe-level p-value	p-value	q-bound	% of CNV Overlap	Gene Symbols	CancerGeneCensu
chr1:157,432,553-157,504,503	q23.2	CN Gain	71,950	17.647	45	-27.353	NaN	1.42E-02	1.0	0	CADM3, LOC100131825, DARC	
chr1:158,627,312-158,915,892	q23.2 - q23.3	CN Gain	288,580	17.647	52.5	-34.853	NaN	3.28E-03	1.0	0	VANGL2, SLAMF6, CD84, SLAMF1, CD4...	
chr1:159,911,585-159,912,192	q23.3	CN Gain	607	26.471	55	-28.529	NaN	1.82E-02	1.0	100	FCGR2B	FCGR2B
chr1:160,003,628-160,016,522	q23.3	CN Gain	12,894	20.588	52.5	-31.912	NaN	7.71E-03	1.0	100	ATF6	
chr1:161,732,835-161,823,417	q23.3	CN Gain	90,582	17.647	52.5	-34.853	NaN	3.28E-03	1.0	0		
chr1:164,155,255-164,334,731	q24.1	CN Gain	179,476	20.588	47.5	-26.912	NaN	2.70E-02	1.0	0	FAM78B	
chr1:164,506,999-164,513,336	q24.1	CN Gain	6,337	20.588	47.5	-26.912	NaN	2.70E-02	1.0	0		
chr3:48,518,015-48,626,543	p21.31	CN Loss	108,528	35.294	7.5	27.794	NaN	3.85E-03	7.11E-01	0	PFKFB4, UCN2, MIR711, COL7A1, UQC...	
chr3:50,814,923-52,749,106	p21.31 - p21.1	CN Loss	1,934,183	35.294	10	25.294	NaN	1.11E-02	7.11E-01	0.698	DOCK3, MANF, RBM15B, VPRBP, RAD54...	BAP1, PBRM1
chr3:53,877,294-54,235,970	p21.1	CN Loss	358,676	35.294	10	25.294	NaN	1.11E-02	7.11E-01	0.045	ACTR8, SELK, CACNA2D3	
chr3:55,107,267-56,432,820	p14.3	CN Loss	1,325,553	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	2.983	WNT5A, ERC2-IT1, MIR3938, ERC2,...	
chr3:59,786,589-			274,095	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	6.171	FHIT	FHIT
chr3:60,064,718-			147,880	41.176	15	26.176	NaN	1.75E-02	7.11E-01	100	FHIT	FHIT
chr6:109,212,67			95,957	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0	ARMC2	
chr6:112,132,07			2,216,091	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.363	FYN, WISP3, TUBE1, FAM229B, LAMA4,...	
chr6:115,247,984-117,551,815	q22.1 - q22.2	CN Loss	2,303,831	41.176	15	26.176	NaN	1.75E-02	7.11E-01	1.721	FRK, TPI1P3, COL10A1, NT5DC1, TSPY...	
chr6:119,982,052-120,034,386	q22.31	CN Loss	52,334	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	0		
chr6:145,035,460-145,602,030	q24.2	CN Loss	566,570	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0	UTRN	
chr6:145,636,936-149,335,506	q24.2 - q25.1	CN Loss	3,698,570	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.352	EPM2A, FBXO30, LOC100507557, SHPR...	
chr6:150,811,437-151,190,460	q25.1	CN Loss	379,023	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	0	PLEKHG1	
chr6:152,281,147-152,449,675	q25.1	CN Loss	168,528	41.176	15	26.176	NaN	1.75E-02	7.11E-01	5.758	ESR1	
chr6:153,124,340-153,625,232	q25.2	CN Loss	500,892	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.211	FBXO5, MTRF1L, RGS17	
chr6:155,467,354-157,120,566	q25.2 - q25.3	CN Loss	1,653,212	41.176	15	26.176	NaN	1.75E-02	7.11E-01	27.457	TIAM2, CLDN20, TFB1M, NOX3,...	
chr6:161,551,021-162,451,528	q26	CN Loss	900,507	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	2.044	AGPAT4, PARK2	
chr6:162,475,389-163,371,481	q26	CN Loss	896,092	41.176	15	26.176	NaN	1.75E-02	7.11E-01	40.582	PARK2, PACRG	
chr6:164,640,702-166,552,368	q27	CN Loss	1,911,666	41.176	15	26.176	NaN	1.75E-02	7.11E-01	6.387	C6orf118, PDE10A, LINC00473, LINC00...	
chr6:166,734,766-167,071,567	q27	CN Loss	336,801	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	0	RPS6KA2-IT1, MIR1913, RPS6KA2	
chr6:168,233,620-168,290,856	q27	CN Loss	57,236	47.059	17.5	29.559	NaN	1.09E-02	7.11E-01	100		
chr10:13,789,296-13,874,667	p13	CN Gain	85,371	41.176	15	26.176	NaN	1.75E-02	1.0	0.54	FRMD4A	
chr13:34,934,183-34,974,239	q13.3	CN Loss	40,056	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	0	NBEA, MAB21L1, MIR548F5	
chr16:15,824,760-15,855,967	p13.11	CN Gain	31,207	0	25	-25	NaN	1.36E-03	1.0	0	MYH11	MYH11
chr16:15,973,870-15,992,796	p13.11	CN Gain	18,926	0	27.5	-27.5	NaN	6.22E-04	1.0	100	ABCC1	

Peaks Only

P-Value Threshold 0.05

Differential Threshold 25.0 %

BioDiscovery



1.28% (686 of 53396) samples

Id	Sample Name	Project Name	Regions	Length	Classification	Notes
2e-01wstwkzhl_000	TCGA-14-1454-01A-01D-051…	TCGA-GBM-Hudson	chr3:46,…	6,584,268		
2e-01wstwkzhl_003	TCGA-14-1459-01A-01D-051…	TCGA-GBM-Hudson	chr3:49,…	3,856,187		
2e-01wstwkzhl_005	TCGA-19-1392-01A-01D-051…	TCGA-GBM-Hudson	chr3:52,…	1,254,878		
2e-01wstwkzhl_012	TCGA-19-0964-01A-01D-051…	TCGA-GBM-Hudson	chr3:40,…	32,292,450		
2e-01wstwkzhl_013	TCGA-14-1452-01A-01D-051…	TCGA-GBM-Hudson	chr3:48,…	4,749,804		
2e-01wstwkzhl_024	TCGA-15-1447-01A-01D-051…	TCGA-GBM-Hudson	chr3:36,…	37,996,793		
2e-01wstwkzhl_026	TCGA-19-0955-01A-02D-051…	TCGA-GBM-Hudson	chr3:43,…	8,086,913		
2e-01wstwkzhl_033	TCGA-26-1440-01A-01D-051…	TCGA-GBM-Hudson	chr3:43,…	11,376,077		
2e-01wstwkzhl_034	TCGA-08-0392-01A-01G-029…	TCGA-GBM-Hudson	chr3:50,…	466,577		
2e-01wstwkzhl_051	TCGA-16-1047-01B-01D-051…	TCGA-GBM-Hudson	chr3:51,…	4,460,095		
2e-01wstwkzhl_074	TCGA-02-0083-01A-01D-019…	TCGA-GBM-Hudson	chr3:0-7…	75,842,787		
2e-01wstwkzhl_080	TCGA-16-0849-01A-01D-038…	TCGA-GBM-Hudson	chr3:30,…	60,183,657		
2e-01wstwkzhl_083	TCGA-02-0114-01A-01D-019…	TCGA-GBM-Hudson	chr3:37,…	48,573,218		
2e-01wstwkzhl_092	TCGA-02-0104-01A-01G-029…	TCGA-GBM-Hudson	chr3:52,…	2,802,964		
2e-01wstwkzhl_096	TCGA-08-0380-01A-01G-029…	TCGA-GBM-Hudson	chr3:52,…	1,271,861		
2e-01wstwkzhl_103	TCGA-06-0745-01A-01D-033…	TCGA-GBM-Hudson	chr3:42,…	48,306,117		
2e-01wstwkzhl_107	TCGA-02-0015-01A-01G-029…	TCGA-GBM-Hudson	chr3:26,…	31,732,337		
2e-01wstwkzhl_111	TCGA-06-0119-01A-08D-021…	TCGA-GBM-Hudson	chr3:51,…	50,774		
2e-01wstwkzhl_131	TCGA-06-0878-01A-01D-038…	TCGA-GBM-Hudson	chr3:15,…	58,086,705		
2e-01wstwkzhl_179	TCGA-02-0113-01A-01D-019…	TCGA-GBM-Hudson	chr3:30,…	25,692,837		
2e-01wstwkzhl_207	TCGA-12-0776-01A-01D-033…	TCGA-GBM-Hudson	chr3:49,…	6,291,938		
2e-01wstwkzhl_224	TCGA-02-0014-01A-01D-018…	TCGA-GBM-Hudson	chr3:31,…	22,268,767		
2e-01wsxmw74n_019	TCGA-02-0014-01A-01D	TCGA-GBM-Stanford	chr3:31,…	22,268,767		
2e-01wsxmw74n_025	TCGA-08-0380-01A-01G-029…	TCGA-GBM-Stanford	chr3:52,…	1,271,861		
2e-01wsxmw74n_037	TCGA-02-0015-01A-01G-029…	TCGA-GBM-Stanford	chr3:26,…	31,732,337		
2e-01wsxmw74n_047	TCGA-08-0392-01A-01G-029…	TCGA-GBM-Stanford	chr3:50,…	466,577		
2e-01wsxmw74n_072	TCGA-02-0104-01A-01G-029…	TCGA-GBM-Stanford	chr3:52,…	2,802,964		
2e-01wsxmw74n_083	TCGA-02-0083-01A-01D	TCGA-GBM-Stanford	chr3:0-7…	75,842,787		
2e-01wsxmw74n_094	TCGA-02-0113-01A-01D	TCGA-GBM-Stanford	chr3:30,…	25,692,837		
2e-01wsxmw74n_139	TCGA-06-0119-01A-08D-021…	TCGA-GBM-Stanford	chr3:51,…	50,774		
2e-01wsxmw74n_156	TCGA-02-0114-01A-01D	TCGA-GBM-Stanford	chr3:37,…	48,573,218		

Query:chr3:50,814,923-52,749,106

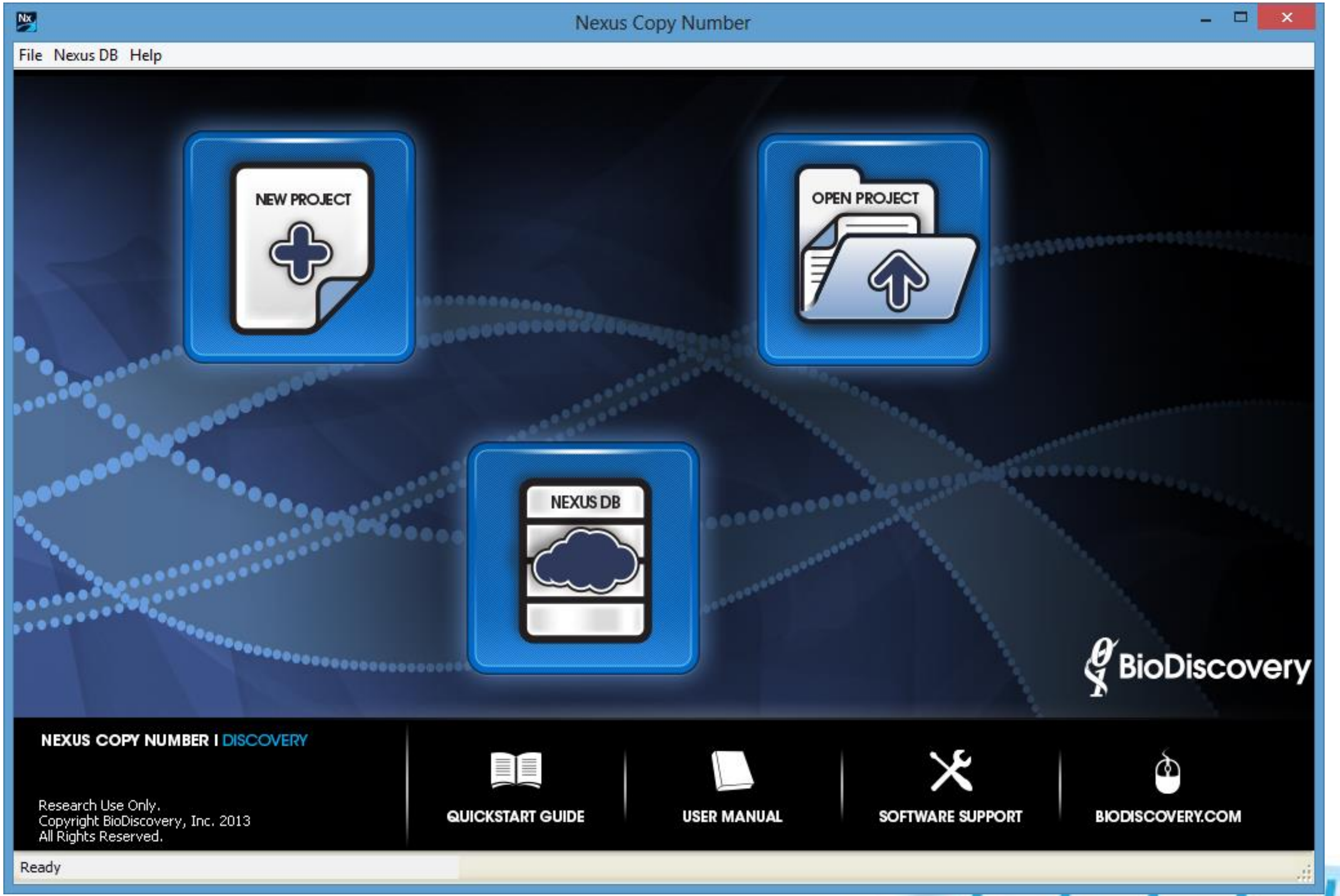
Results range:chr3:0-199,298,372

View:chr3:0-199,298,372

What if I have my own data?

Project Set-up and Processing

Launch Nexus Copy Number


Nexus Copy Number
File Nexus DB Help
NEW PROJECT
OPEN PROJECT
NEXUS DB
BioDiscovery
NEXUS COPY NUMBER I DISCOVERY
QUICKSTART GUIDE
USER MANUAL
SOFTWARE SUPPORT
BIODISCOVERY.COM
Ready

Research Use Only.
Copyright BioDiscovery, Inc. 2013
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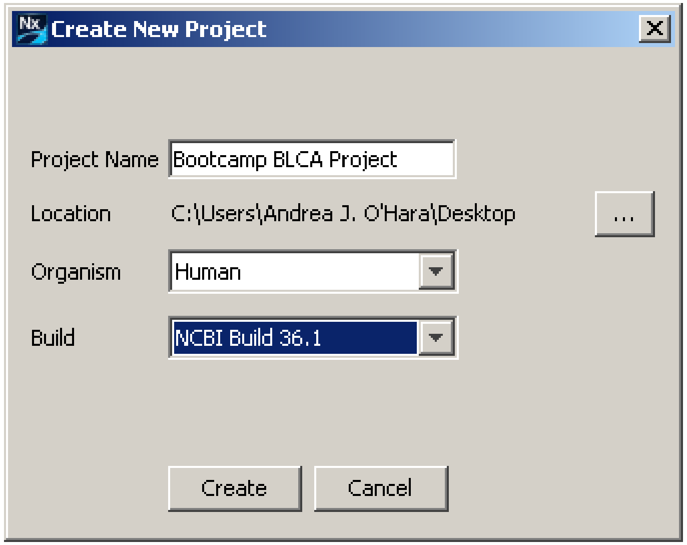
Creating A New Project

- Specify project name
- Select location to save the project
- Specify organism (Human) and build (NCBI Build 36.1)
- Click ‘Create’


Create New Project

Project Name: Bootcamp BLCA Project

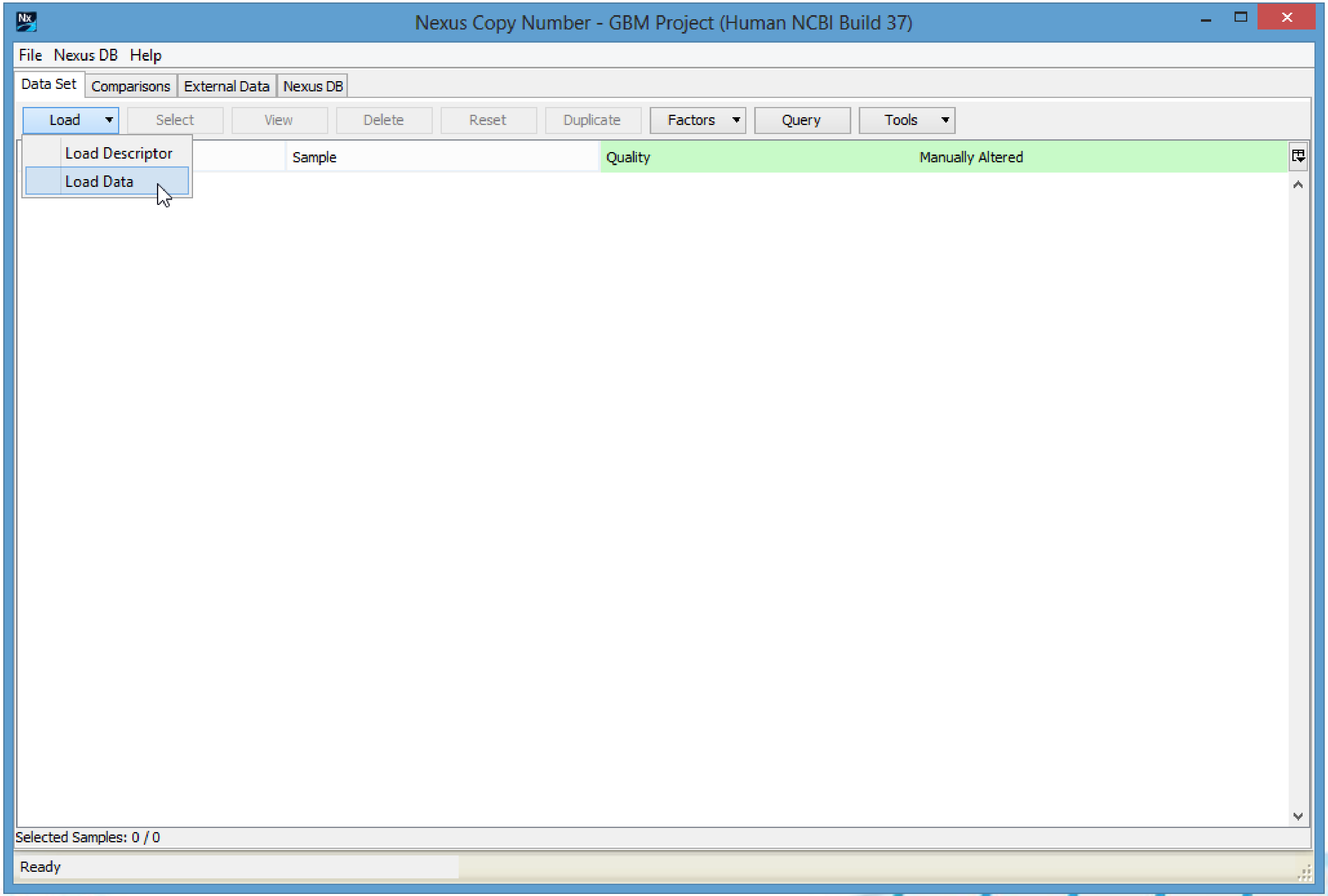
Location: C:\Users\Andrea J. O'Hara\Desktop

Organism: Human

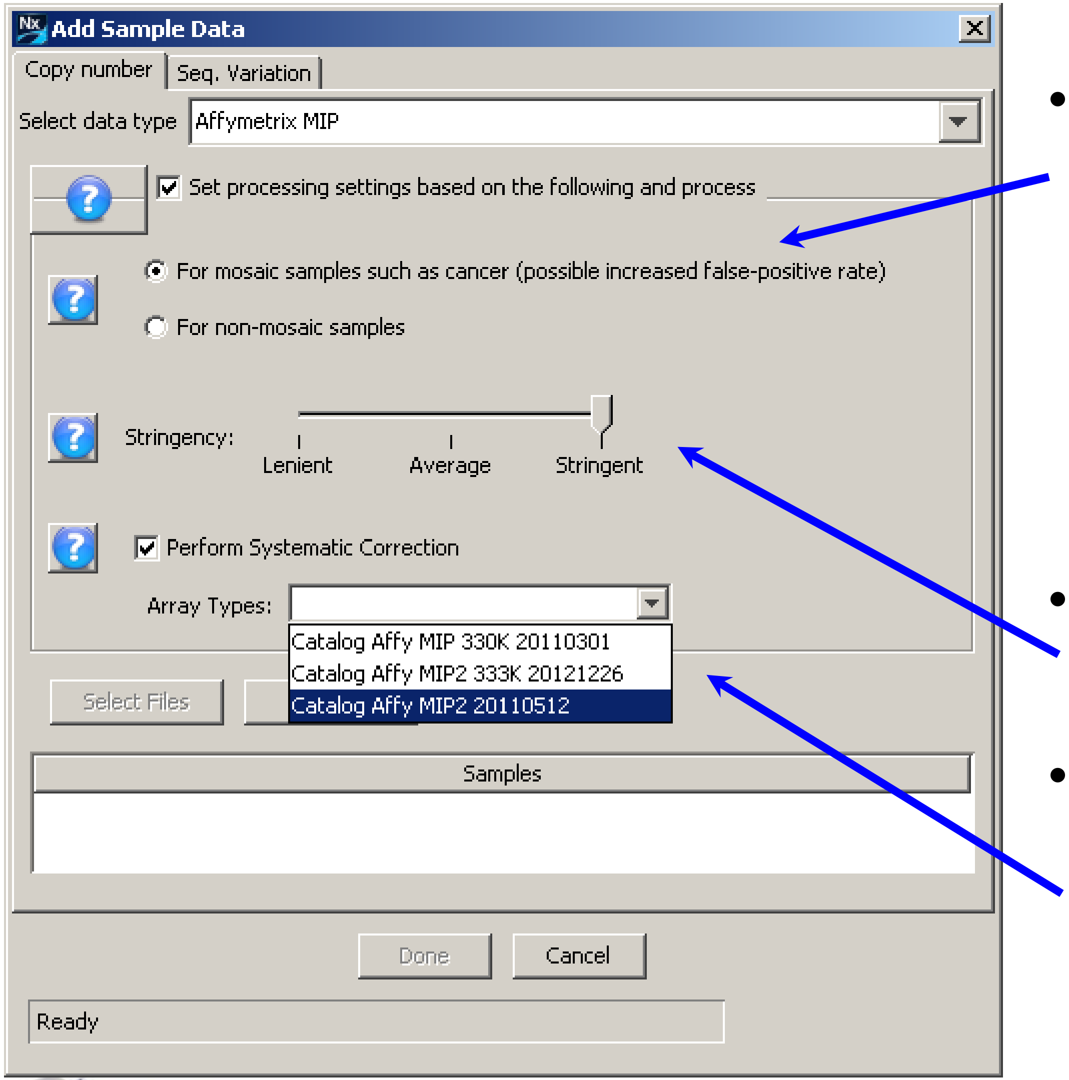
Build: NCBI Build 36.1

...
Create
Cancel

Load Data


Nexus Copy Number - GBM Project (Human NCBI Build 37)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Nexus DB
Load
Select
View
Delete
Reset
Duplicate
Factors
Query
Tools
Load Descriptor
Load Data
Sample
Quality
Manually Altered
Selected Samples: 0 / 0
Ready

Parameters Affected by the Load Data Guide



Select data type: Affymetrix MIP

☑ Set processing settings based on the following and process

● For mosaic samples such as cancer (possible increased false-positive rate)

○ For non-mosaic samples

Stringency: Lenient — Average — Stringent (set to Stringent)

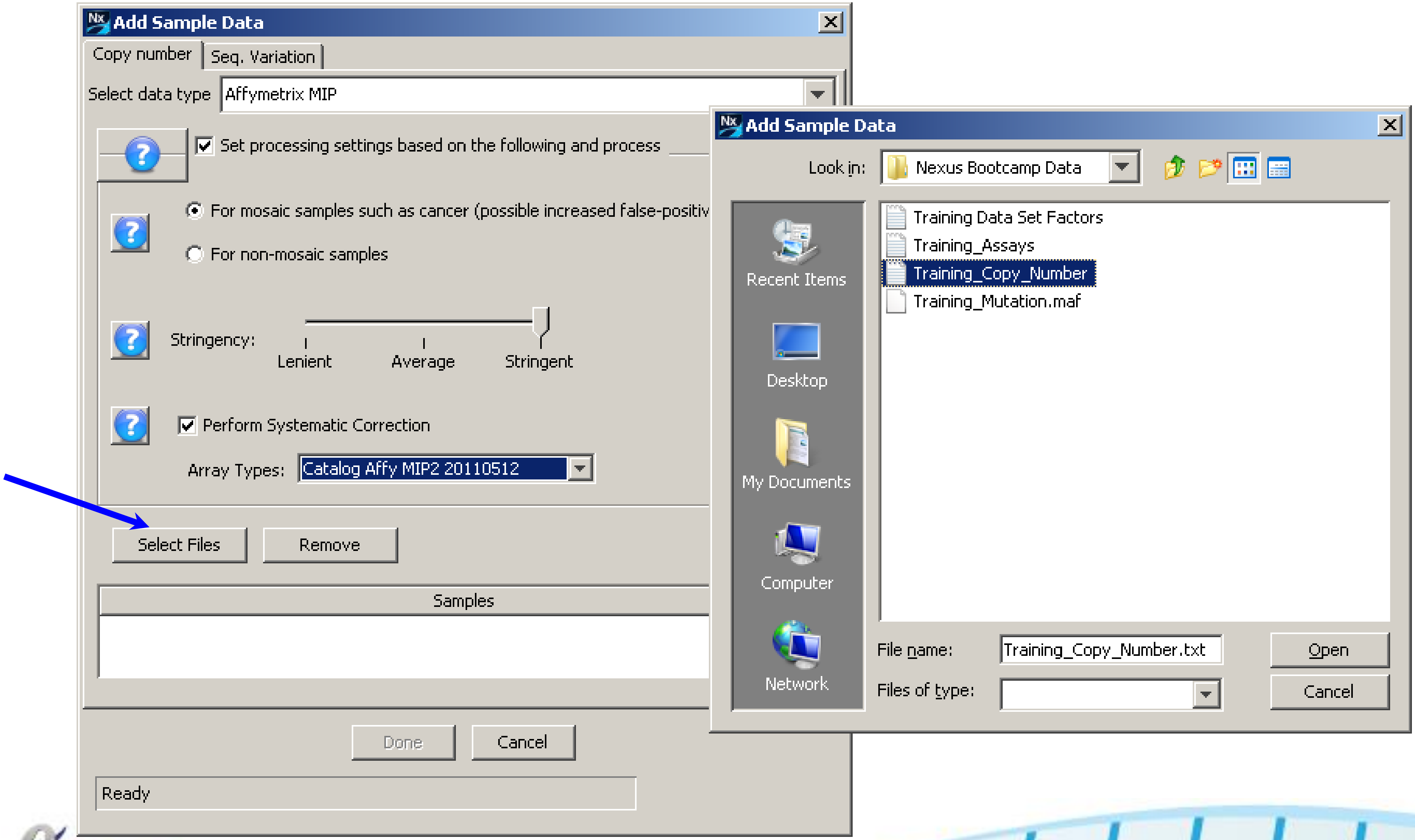
☑ Perform Systematic Correction

Array Types:
- Catalog Affy MIP 330K 20110301
- Catalog Affy MIP2 333K 20121226
- Catalog Affy MIP2 20110512

Ready

- Adjusts call thresholds (e.g. if have mix of cancer and normal samples, thresholds are lowered from what one would expect theoretically)
- Stringency adjusts the significance threshold
- Select array type for loading appropriate correction file for systematic correction

Import Data File



Add Sample Data

Tabs: Copy number | Seq. Variation

Select data type: Affymetrix MIP

☑ Set processing settings based on the following and process

● For mosaic samples such as cancer (possible increased false-positiv

○ For non-mosaic samples

Stringency: Stringent

☑ Perform Systematic Correction

Array Types: Catalog Affy MIP2 20110512

Ready

Add Sample Data

Look in: Nexus Bootcamp Data

- Training Data Set Factors
- Training_Assays
- Training_Copy_Number
- Training_Mutation.maf

File name: Training_Copy_Number.txt

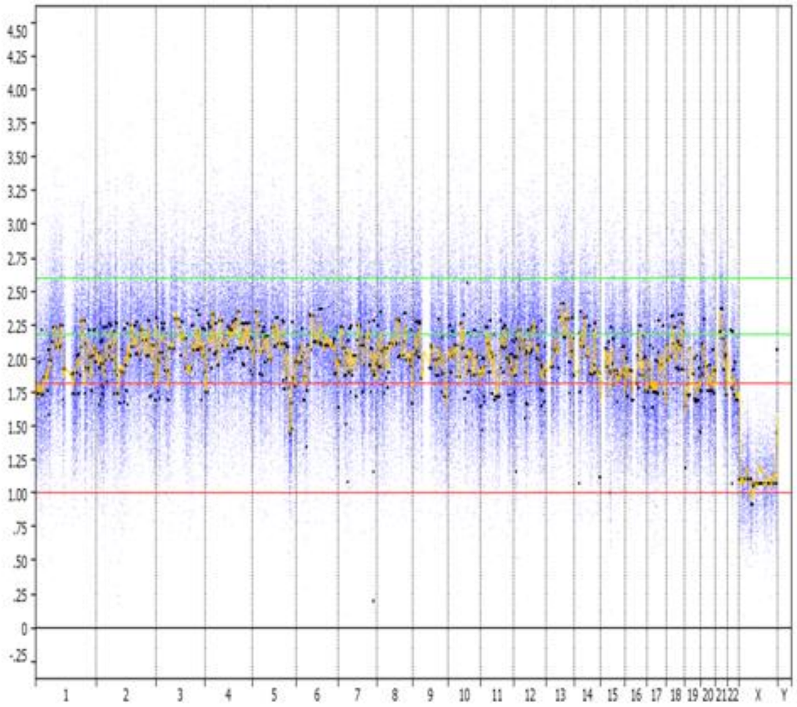
Files of type:

Lenient
Average
Stringent
Select Files
Remove
Samples
Done
Cancel
Recent Items
Desktop
My Documents
Computer
Network
Open
Cancel

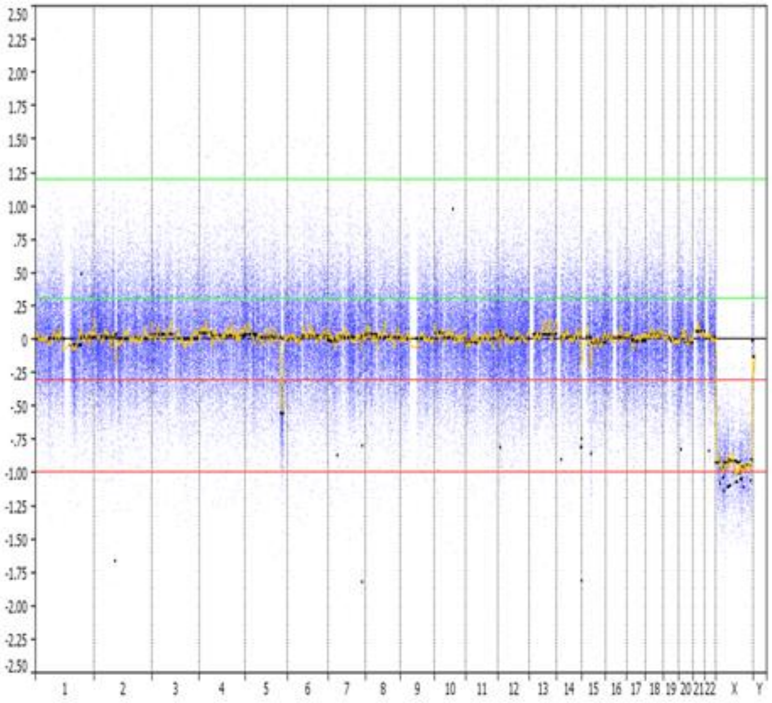
Pre-processing of Data in Nexus

- Systematic correction

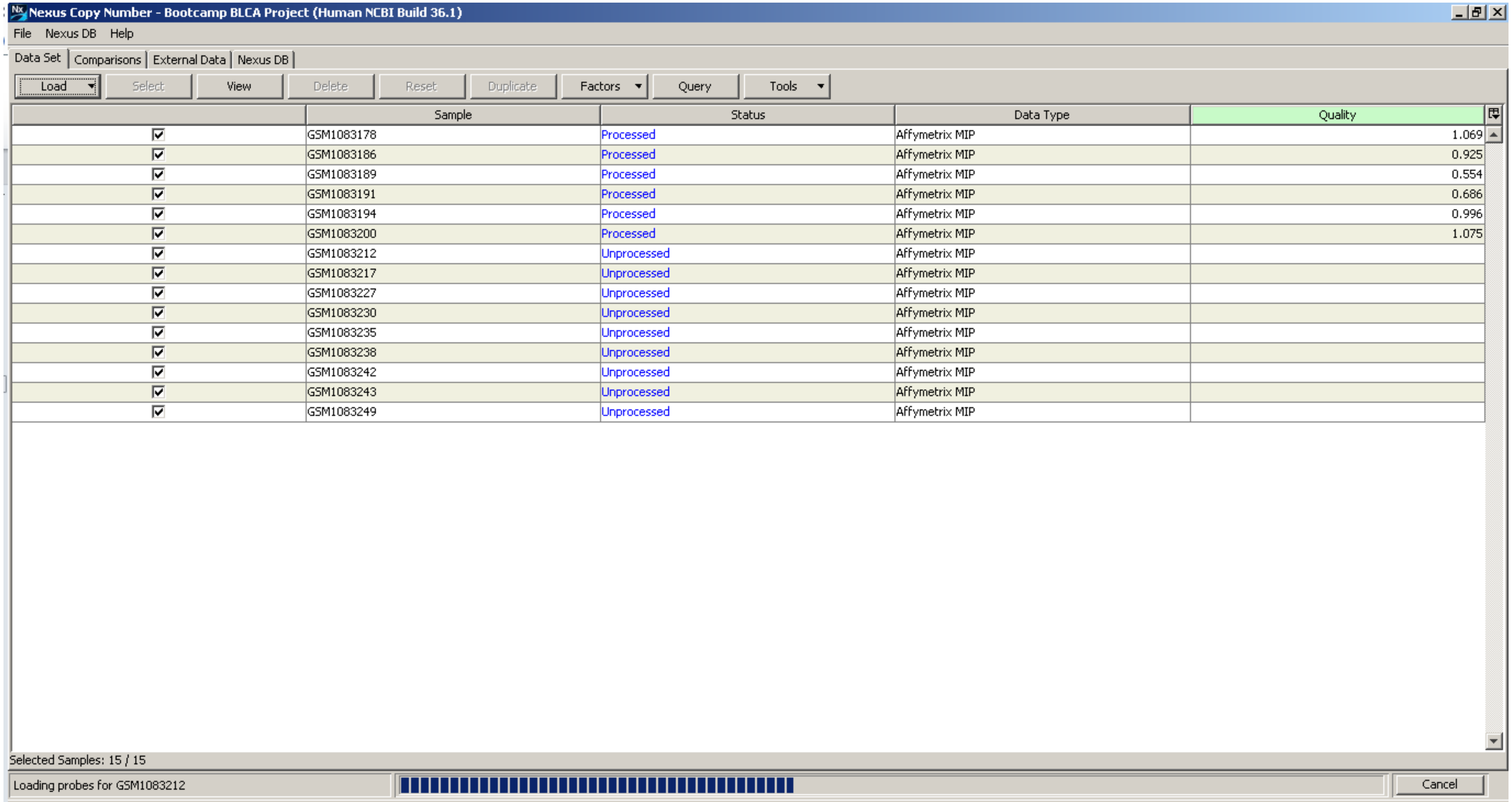
Before



After



Process Samples

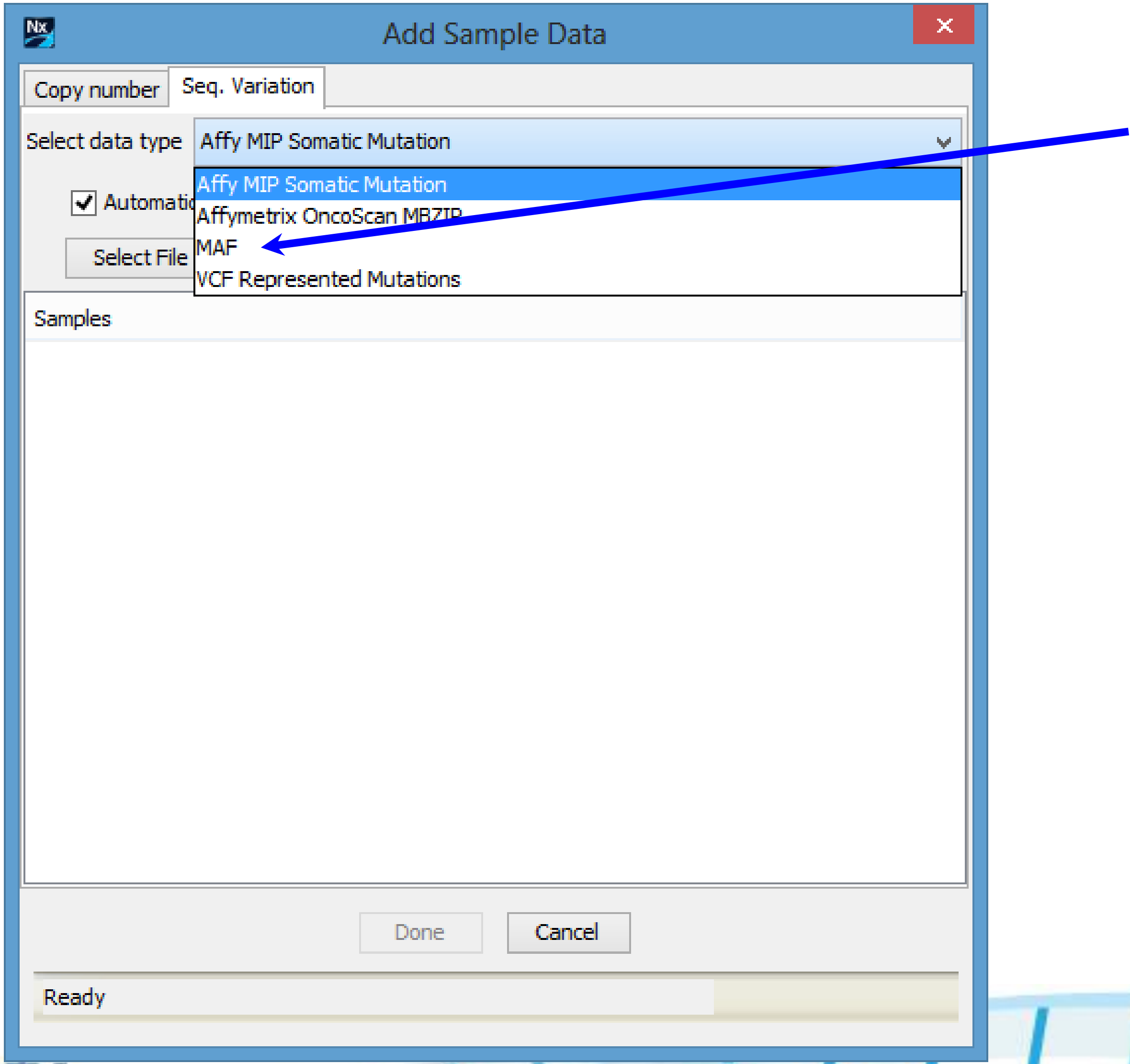

Nexus Copy Number - Bootcamp BLCA Project (Human NCBI Build 36.1)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Nexus DB
Load
Select
View
Delete
Reset
Duplicate
Factors
Query
Tools
Cancel

	Sample	Status	Data Type	Quality
☑	GSM1083178	Processed	Affymetrix MIP	1.069
☑	GSM1083186	Processed	Affymetrix MIP	0.925
☑	GSM1083189	Processed	Affymetrix MIP	0.554
☑	GSM1083191	Processed	Affymetrix MIP	0.686
☑	GSM1083194	Processed	Affymetrix MIP	0.996
☑	GSM1083200	Processed	Affymetrix MIP	1.075
☑	GSM1083212	Unprocessed	Affymetrix MIP	
☑	GSM1083217	Unprocessed	Affymetrix MIP	
☑	GSM1083227	Unprocessed	Affymetrix MIP	
☑	GSM1083230	Unprocessed	Affymetrix MIP	
☑	GSM1083235	Unprocessed	Affymetrix MIP	
☑	GSM1083238	Unprocessed	Affymetrix MIP	
☑	GSM1083242	Unprocessed	Affymetrix MIP	
☑	GSM1083243	Unprocessed	Affymetrix MIP	
☑	GSM1083249	Unprocessed	Affymetrix MIP	

Selected Samples: 15 / 15

Loading probes for GSM1083212

Import Sequence Variants Data

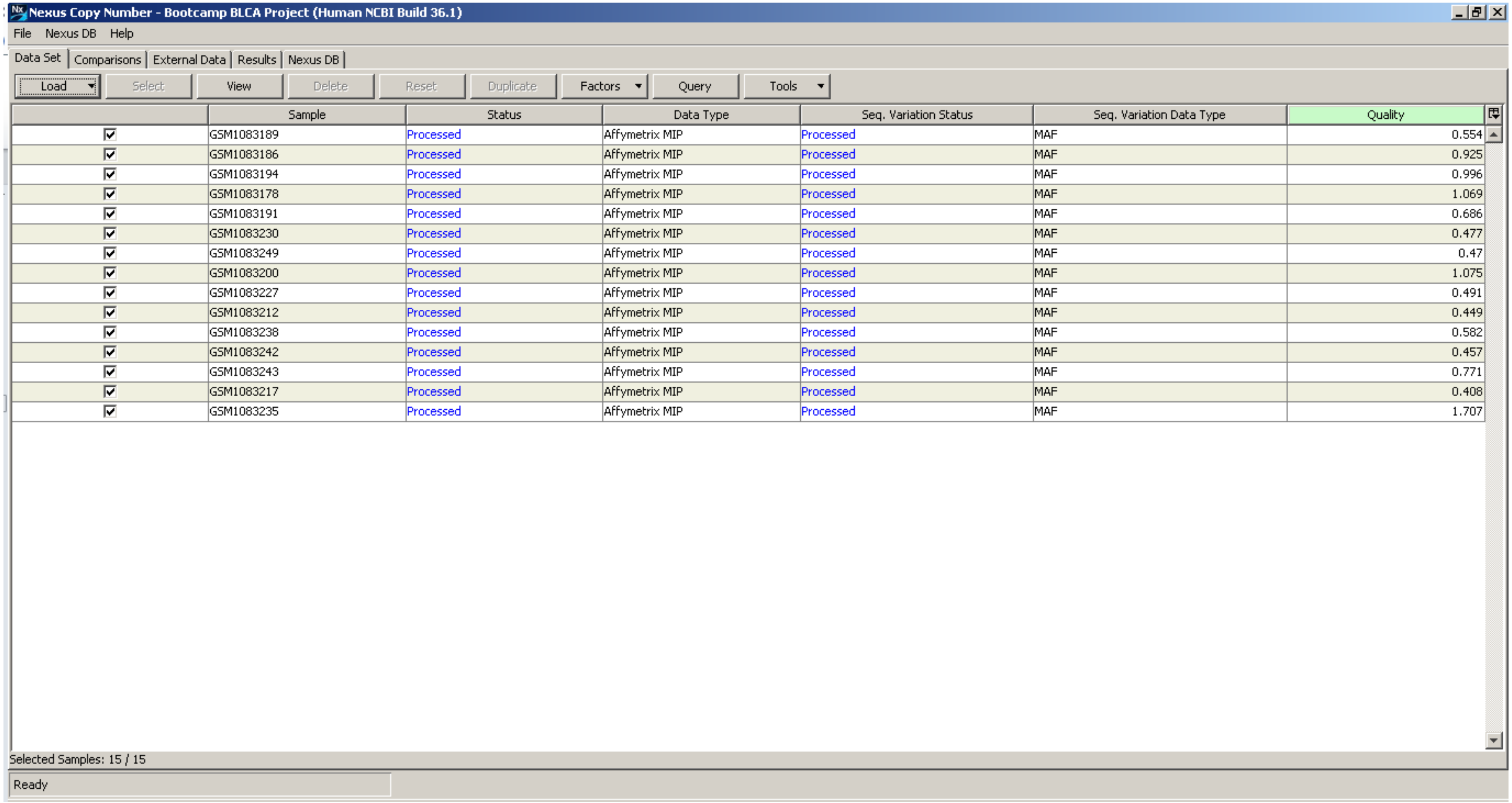

Add Sample Data
Copy number
Seq. Variation
Automatic
Select File
Samples
Done
Cancel

Select data type: Affy MIP Somatic Mutation

- Affy MIP Somatic Mutation
- Affymetrix OncoScan MBZIP
- MAF
- VCF Represented Mutations

Ready

Import Sequence Variants Data


Nexus Copy Number - Bootcamp BLCA Project (Human NCBI Build 36.1)
File
Nexus DB
Help
Data Set
Comparisons
External Data
Results
Nexus DB
Load
Select
View
Delete
Reset
Duplicate
Factors
Query
Tools

	Sample	Status	Data Type	Seq. Variation Status	Seq. Variation Data Type	Quality
☑	GSM1083189	Processed	Affymetrix MIP	Processed	MAF	0.554
☑	GSM1083186	Processed	Affymetrix MIP	Processed	MAF	0.925
☑	GSM1083194	Processed	Affymetrix MIP	Processed	MAF	0.996
☑	GSM1083178	Processed	Affymetrix MIP	Processed	MAF	1.069
☑	GSM1083191	Processed	Affymetrix MIP	Processed	MAF	0.686
☑	GSM1083230	Processed	Affymetrix MIP	Processed	MAF	0.477
☑	GSM1083249	Processed	Affymetrix MIP	Processed	MAF	0.47
☑	GSM1083200	Processed	Affymetrix MIP	Processed	MAF	1.075
☑	GSM1083227	Processed	Affymetrix MIP	Processed	MAF	0.491
☑	GSM1083212	Processed	Affymetrix MIP	Processed	MAF	0.449
☑	GSM1083238	Processed	Affymetrix MIP	Processed	MAF	0.582
☑	GSM1083242	Processed	Affymetrix MIP	Processed	MAF	0.457
☑	GSM1083243	Processed	Affymetrix MIP	Processed	MAF	0.771
☑	GSM1083217	Processed	Affymetrix MIP	Processed	MAF	0.408
☑	GSM1083235	Processed	Affymetrix MIP	Processed	MAF	1.707

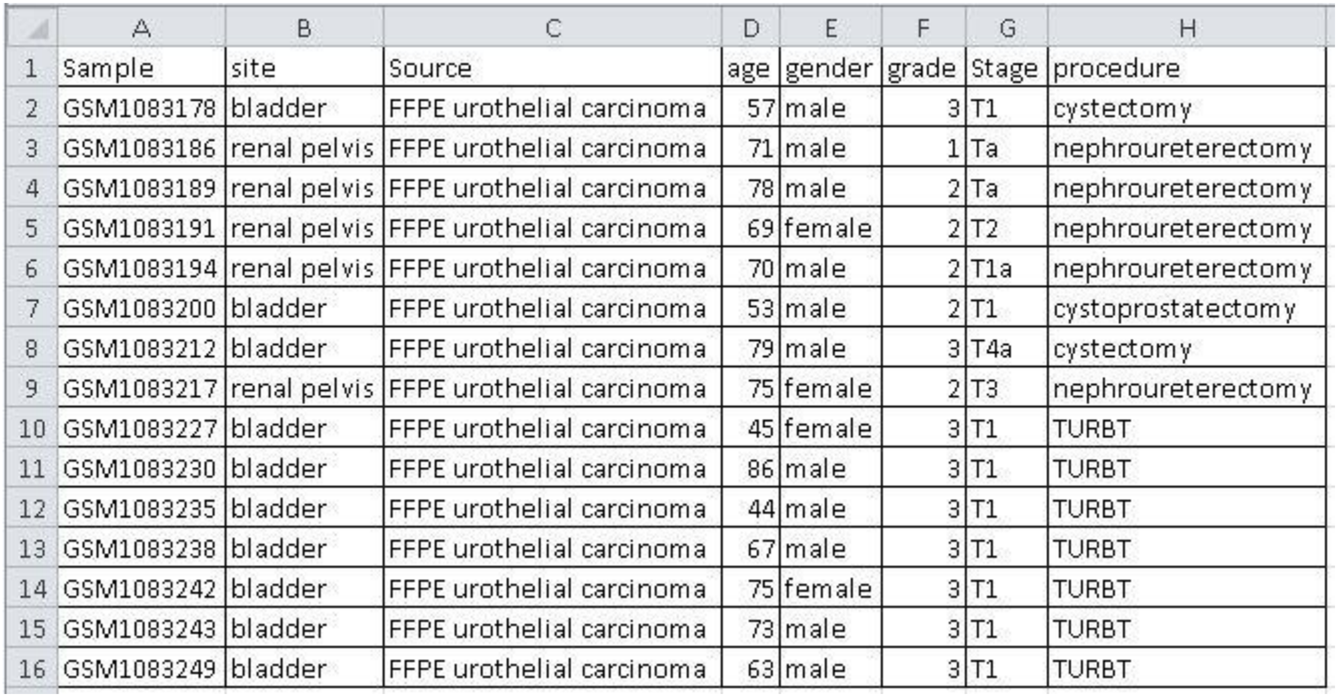
Selected Samples: 15 / 15

Ready

Data Set Tab – Factors

- Any clinical data or phenotypic data associated with a sample, e.g. gender, age, tumor type, tumor grade
- Each sample can have an unlimited number of Factors
- Optional but important
- Can be entered manually or loaded from a tab-delimited text file in the Data Set tab

Load Factors – Example Factors File



	A	B	C	D	E	F	G	H
1	Sample	site	Source	age	gender	grade	Stage	procedure
2	GSM1083178	bladder	FFPE urothelial carcinoma	57	male	3	T1	cystectomy
3	GSM1083186	renal pelvis	FFPE urothelial carcinoma	71	male	1	Ta	nephroureterectomy
4	GSM1083189	renal pelvis	FFPE urothelial carcinoma	78	male	2	Ta	nephroureterectomy
5	GSM1083191	renal pelvis	FFPE urothelial carcinoma	69	female	2	T2	nephroureterectomy
6	GSM1083194	renal pelvis	FFPE urothelial carcinoma	70	male	2	T1a	nephroureterectomy
7	GSM1083200	bladder	FFPE urothelial carcinoma	53	male	2	T1	cystoprostatectomy
8	GSM1083212	bladder	FFPE urothelial carcinoma	79	male	3	T4a	cystectomy
9	GSM1083217	renal pelvis	FFPE urothelial carcinoma	75	female	2	T3	nephroureterectomy
10	GSM1083227	bladder	FFPE urothelial carcinoma	45	female	3	T1	TURBT
11	GSM1083230	bladder	FFPE urothelial carcinoma	86	male	3	T1	TURBT
12	GSM1083235	bladder	FFPE urothelial carcinoma	44	male	3	T1	TURBT
13	GSM1083238	bladder	FFPE urothelial carcinoma	67	male	3	T1	TURBT
14	GSM1083242	bladder	FFPE urothelial carcinoma	75	female	3	T1	TURBT
15	GSM1083243	bladder	FFPE urothelial carcinoma	73	male	3	T1	TURBT
16	GSM1083249	bladder	FFPE urothelial carcinoma	63	male	3	T1	TURBT

Additional Information

Webinars Library:

http://www.biodiscovery.com/video-library/webinars/

Educational Video Library:

http://www.biodiscovery.com/video-library/educational-videos/

Blog Postings:

http://www.blog.biodiscovery.com/

White Papers:

http://www.biodiscovery.com/white-papers/

Additional Information

Nexus Copy Number (TCGA Copy Number and Mutation):

http://www.biodiscovery.com/copy_number_cf.php

Nexus Expression (TCGA RNA-Seq and Expression Arrays):

http://www.biodiscovery.com/expression_cf.php

Please put in a ticket with the help desk to complete installation and gain access to the license server