BioDiscovery Nexus Training Course

### **Nexus Copy Number Training**

Andrea O'Hara, Ph.D.

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



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### **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



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### **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

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### **Nexus Copy Number Workflow**





### Segmenting the Probes



### **B-Allele Freq. Bands**





### 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

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### The Cancer Genome Atlas (TCGA)

### 26 Tumor Types (+6)



### 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:

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- 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 - Esop	hageal Cancer Pi	roject (Human NC	BI Build 36.1)	- 🗇 🗙						
File Nexus DB Help										
Data Set Comparisons External Data Results Nexus DB										
		NX	Register Account							
🖉 Login 🗙		First Name	John							
		Last Name	Doe							
Username		Email	jdoe@mdacc.org							
Password NDB		Domain	NDB							
Ok Cancel	Log in	Address								
Register Account Forgot Password		Phone								
			Submit Cancel							
User name: tcga_training Password: TCGAtraining14! <u>http://chilp.it/0205</u>										
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### Nexus DB Data Download

<b>N</b>	Ne	xus Copy Number - Es	ophageal Cancer Pro	oject (Human NCBI B	uild 36.1)			_ 0	x
File Nexus DB Hel	p								
Data Set Compariso	ns External Data Results Nexus DB								
									_
My Projects	Visible Projects My Groups Change Password								
Select	Refresh Filter Download								
			1						_
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	3
	TCGA-OV-Broad	TCGA OV samples run at t	tumor, OV, TCGA, Affy	429	Human NCBI Build 36.1	Affymetrix	craig	TCGA,BDI Employees	3
	TCGA-GBM-Stanford	TGCA data for GBM run o	GBM, cancer, Illumina, H	157	Human NCBI Build 36.1	Illumina	craig	TCGA,BDI Employees	3
	TCGA-GBM-Hudson	TCGA GBM Tumor samples	. TCGA,Human,cancer,	270	Human NCBI Build 36.1	Illumina	craig	TCGA,BDI Employees	3
	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	2
	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	Affvmetrix.Level 3	raia	BDI Employees.Public	

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Ready

### Nexus DB Data Download

		Select	Download Downloa	d & View Email	Samples :605 / Selected: 6			
	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			
•	001	TCGA_Level3_seg	TCGA-18-3406-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCO		
•	002	TCGA_Level3_seg	TCGA-18-3407-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	тсе		
<	003	TCGA_Level3_seg	TCGA-18-3407-01A-01D-1969-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCO		
<	004	TCGA_Level3_seg	TCGA-18-3407-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
•	005	TCGA_Level3_seg	TCGA-18-3408-01A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	тсе		
•	006	TCGA_Level3_seg	TCGA-18-3408-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	тсе		
•	007	TCGA_Level3_seg	TCGA-18-3409-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	тсо		
•	008	TCGA_Level3_seg	TCGA-18-3409-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	тсо		
<	009	TCGA_Level3_seg	TCGA-18-3410-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCO		
<	010	TCGA_Level3_seg	TCGA-18-3410-01A-01D-1969-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	тсо		
<	011	TCGA_Level3_seg	TCGA-18-3410-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCO		
~	012	TCGA_Level3_seg	TCGA-18-3411-01A-01D-0978-01	Current smoker	Lung Squamous Cell Carcinoma	тсо		
•	013	TCGA_Level3_seg	TCGA-18-3411-01A-01D-1969-01	Current smoker	Lung Squamous Cell Carcinoma	тсо		
•	014	TCGA_Level3_seg	TCGA-18-3411-11A-01D-0978-01	Current smoker	Lung Squamous Cell Carcinoma	тсе		
•	015	TCGA_Level3_seg	TCGA-18-3412-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCO		
•	016	TCGA_Level3_seg	TCGA-18-3412-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
~	017	TCGA_Level3_seg	TCGA-18-3414-01A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCG		
•	018	TCGA_Level3_seg	TCGA-18-3414-11A-01D-0978-01	Current reformed smoker for < or = 15 years	Lung Squamous Cell Carcinoma	TCG		
•	019	TCGA_Level3_seg	TCGA-18-3415-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
•	020	TCGA_Level3_seg	TCGA-18-3415-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
<	021	TCGA_Level3_seg	TCGA-18-3416-01A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
•	022	TCGA_Level3_seg	TCGA-18-3416-11A-01D-0978-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		
~	023	TCGA_Level3_seg	TCGA-18-3417-01A-01D-1439-01	Current reformed smoker for > 15 years	Lung Squamous Cell Carcinoma	TCG		

### 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.



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



### **Downstream Analysis**



### **Results – Genome View**



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### Single Sample Drill Down – Sample Info

CGA-KL-8340-01A-11D-23 ple Info Overview Whole Ge	08-01_Paired (2e-023k9bpmmk_25) - 5a enome   Chromosome   Summary	ample Drill Down 📃 🗖
	File	Direction
mple	I:\TCGA\Raw Data\KICH\SNP\.\BI F	Forward
ample	I:\TCGA\Raw Data\KICH\SNP\.\BI F	Forward
Sample Name	D-2308-01_Paired (2e-023k9bpmmk_25)	
Status	Processed	
Seq. Variation Status	Processed	
Quality	0.08	3
% Genome Changed	31.44	4
One copy gain		
Two or more copy gain		ז
One copy loss	29	P
Two copy loss		ז
Total CN aberrations	29	Ð
Manually Altered		
LOH		ז
% LOH	0.00	0
Discarded %	0.13	3
Point Mutations	7	7
Insertions	(	
Deletions	1	

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### Single Sample Drill Down – Overview



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### Single Sample Drill Down – Whole Genome



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### Single Sample Drill Down – Chromosome



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### Mark Location 8 Horizontal Zoom 16 Rectangular Zoom Q +Ô+ Center View Locate Region Query Database Eð **P** Region Query Settings Filter

Add/Join Region



### Single Sample Drill Down – Modify View

	<b>N</b>	Sele	ct		×	
San					^	05810 - Sample Drill Down – 🗗 🗙
Chr	Max Size				_	Find
	Max Region					
	Locus IDs					q14.3 q15 q23.1 q23.2 q32 q34
Z	B/P Genes					
E	Call PValue					
n	Notes					
	Classification					and the state of the
	Normal					
					-1	
	Counts	IDs				80 Mb 90 Mb 100 Mb 110 Mb 120 Mb 130 Mb 140 Mb 150 Mb 160 Mb 170 Mb 180 Mb
	Gene Symbols 🔽	✓				
	miRNAs					
11		Counts	Elanking	IDs	_	
	Affy 500k					
	Affy CytoScan-HD CNP					
Ir	Affy CytoScan-HD SNP					% of CNV Overlap         Probe Median         % Heterozygous         Probes         Count of Gene Symbols           5.13         0.02         5.31         245         29
	Affy SNP6					100.00 1.15 N/A 24 0 V
	Affy SNP6 CNP					gions Modify View Export TXT # of conjunct 254
Rea	Affy SNP6 SNP					+ 01 regions: 234
Red					~	
	<				>	
2		Apply	Cancel			
	BIODISCO	over	V	-	1	
1			-	1		
						BioDiscovery © 2013

### Single Sample Drill Down – Summary

### TCGA-KL-8340-01A-11D-2308-01\_Paired (2e-023k9bpmmk\_25) - Sample Drill Down \_ 8 × Sample Info | Overview | Whole Genome | Chromosome | Summary 4 chr1 chr2 chr3 chr4 chr5 chr6 chr7 chr8 11 X chr10 chr11 chr12 chr13 chr14 chr15 chr16 X chr17 chr18 chr19 chr20 chr21 chr22 chrX chrY X Ready



## Identifying Frequent and Significant Peaks

- What are "common" aberrations in this set
- What are "significant" common abberrations

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### Aggregate Table Approach

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



### **Aggregate Table**

Data Set Comparisons	External Data Results	Nexus DB												
Genome Chrom	osome <u>Summary</u>	<u>Aggregate</u>												
Export TXT	View Annotations	Participation	Enrichment	õignificant Peaks	Modify Vi	ew								
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		385-11, 1,275,185 p11.2 - p11.1		885-11, 1,275,185 p11.2 - p11.1 CN Loss		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-Valu	e cut-off 0.0		e % cut-off 35.0	D Peaks only							

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### Significance Testing (STAC)



### Significant Peaks (STAC)

Nexus Copy Numbe	Nexus Copy Number - NCI Training Session (Human NCBI Build 37)											
File Nexus DB Help												
Data Set Comparisons	External Data Results	Nexus DB										
Genome Chrom	osome <u>Summary</u>	<u>Aggregate</u>										
Export TXT	View Annotations	Participation	Enrichment	ficant Peaks 🔪 🛛 Mod	ify View							
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 cu	t-off 0.05 Aggr	egate % cut-off 35.1	D Peaks only						

Ready

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### 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)											
Jata Set Comparisons External Data Results Nexus DB											
Genome Chromosome Summary Aggregate											
riew Factors 🛅 🔄 🖑 🕌 🗣 🚱 😰 💱 😓 💂 Search: Find Table Export BGR Enrichments Cluster											
1 2 3 4 5 6 7 8 9 10 11 12 13 14 15 16 1 <sup>°</sup>	7 18 19 20 21 22 X Y										
50%											
CNVs											
TCGA-KM-8439-01A											
ICGA-KM-8441-01A	• • •										
TCGA-KN-8437-01A-*	• •										
TCGA-KL-8326-01A-	••••										
	• • •										
TCGA-KL-8340-01A											
	·										

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### GISTIC



 Specify G-score (combination score of frequency and amplitude) and Q-bound (significance)

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• Only multiple testing correction value calculated, may need to reduce threshold to best capture results (0.25)

### GISTIC

### 🚾 Nexus Copy Number - NCI Training Session (Human NCBI Build 37) \_ 8 × File Nexus DB Help Data Set Comparisons External Data Results Nexus DB Modify View Load View Factors Query Tools • GISTIC % LOH % Genome C Number of Patient Samples 🖽 Sample Status Data Type Seq. Variation Status Seq. Variation Data Type Quality Total CN aberrations id Regions Event Gender TCGA-KL-83... Processed Database Pr... Processed 0.0902 111 3.4924 Concordance function chr4,ch.. Database Processing T... 1 🔺 2 Filter Samples TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0710 88 0.0 chr11 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0852 33 0.0498 Export TXT chr4,ch... 2 22 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0729 0.0 4.6455 1 2 0.1204 52 18.8160 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 57.8643 Low Level Mo... chr1.chr10 - 1 $\mathbf{\nabla}$ TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0758 117 0.0 41.0755 chr3,chr12 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T.. 0.1336 152 0.0 37.5844 chr3.chr7 2 TCGA-KL-83... Processed 0.1291 48 0.0 43.3301 chr3:0-1980.. Database Pr... Processed Database Processing T... 1 2 TCGA-KL-83... Processed Database Processing T... 0.0804 220 0.0220 37.6850 chr3,chr4,ch... Database Pr... Processed 1 V 208 39.4821 chr3.chr4.ch.. TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0970 0.0 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0829 65 0.0 20.4219 Low Level Mo... chr4,chr5 2 54 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0690 0.0 36,3383 chr3,chr11 - 1 2 TCGA-KL-83... Processed 0.0999 112 0.0 34.1472 chr4,chr8 Database Pr... Processed Database Processing T... 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0676 75 0.0 44.6466 chr3,chr4,chr8 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0672 37 0.0 32.4886 chr3.chr11 1---2 52 TCGA-KL-83... Processed Database Processing T... 0.0797 0.0 30.2557 chr3,chr11 1 Database Pr... Processed 7 Database Processing T... 218 2.9705 34.0153 chr7,chr12 TCGA-KI -83... Processed Database Pr... Processed 0.1061 -18 2 0.0763 29 31.4417 chr3,chr8 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.1098 217 0.1348 37.8524 chr4,chr11 1 V 55 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0515 0.0 32,3343 chr3,chr11 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.1023 45 0.0 25.0021 Low Level Mo.. 1 7 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.1523 27 0.0 12.2009 Low Level Mo... 1 2 TCGA-KL-83... Processed Database Pr... Processed Database Processing T... 0.0985 100 0.0 32.3757 chr3, chr8 1 2 55 TCGA-KL-83... Processed Database Processing T... 0.0840 0.1205 44.2263 chr3:140650.. 1 Database Pr... Processed TCGA-KM-8... Processed 2 0.1323 65 0.0183 38,4868 chr4.chr8 Database Pr... Processed Database Processing T... 1 2 TCGA-KM-8... Processed Database Pr... Processed Database Processing T... 0.0763 88 0.0 31.1763 chr3,chr8 1 2 TCGA-KM-8... Processed Database Pr... Processed Database Processing T... 0.0916 1 0.0 0.0240 1 V 0.1023 31 Database Processing T... 0.0 36,0662 cbr3.cbr11 TCGA-KM-8... Processed Database Pr... Processed 1 2 TCGA-KM-8... Processed Database Pr... Processed Database Processing T... 0.0636 148 0.0189 36.1235 chr3,chr12 1 2 0.0733 97 TCGA-KM-8... Processed Database Pr... Processed Database Processing T... 0.0 36.9364 chr3,chr12 1 P 7 TCGA-KM-8... Processed Database Pr... Processed Database Processing T... 0.0997 ol 0.0 0.0 Selected Samples: 65 / 65

Ready

**Start** 

**BioDiscovery** 

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### 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

covery

• Added weight for high copy gains and homozygous deletions

### **GISTIC** Results

Magaza Profile					
Regions Genome Chromosom	ne				
Export TXT Annota	tions				
Region Extended Region		Туре	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

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core cut-off 1.0

### **GISTIC – Genome View**



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### 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.

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# 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

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### Concordance – Chr9p Loss

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

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### Data Set Comparisons External Data Results Nexus DB

File Nexus DB Help

Load	• Select	Vi	ew	Delete	Reset Duplica	te Fa	ctors • Modify	/iew	Query T	ools 💌				
	Sample	Status	Data Type	Seq. Variation Status	Seq. Variation Data Type	Quality	Total CN aberrations	% LOH	% Genome Changeo	Curator's Notes	Diploid Regions	Event	Gender	Number of Patient Samples 🛱
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0902	111	3.4924	35.8427	/	chr3,chr4,ch			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0710	88	0.0	48.4609	9	chr4,chr11			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0852	33	0.0498	32,3733	8	chr3,chr4,ch			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0729	22	0.0	4.6455	5				1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1204	52	18.8160	57,8643	Low Level Mo	chr1,chr10			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0758	117	0.0	41.0755	5	chr3,chr12			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1336	152	0.0	37.5844	ł	chr3,chr7			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.1291	48	0.0	43.3301		chr3:0-1980			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0804	220	0.0220	37,6850	)	chr3,chr4,ch			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0970	208	0.0	39,4821		chr3,chr4,ch			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0829	65	0.0	20.4219	Low Level Mo	chr4,chr5			1
<b>N</b>	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0690	54	0.0	36.3383	3	chr3,chr11			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0999	112	0.0	34.1472	2	chr4,chr8			1
<b>N</b>	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0676	75	0.0	44.6466	5	chr3,chr4,chr8			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0672	37	0.0	32.4886	5	chr3,chr11			11
<b>N</b>	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0797	52	0.0	30.2557	/	chr3,chr11			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1061	218	2.9705	34.0153	3	chr7,chr12			1
<b>N</b>	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0763	29	0.0	31.4417	/	chr3,chr8			1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1098	217	0.1348	37.852		abed abel 1			1
<b>N</b>	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0515	55	0.0	32.3	👺 Query P	Panel		X	1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1023	45	0.0	25.0					1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1523	27	0.0	12.2	Term:	Region		<b>•</b>	1
	TCGA-KL-83 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0985	100	0.0	32.3		1			1
	TCGA-KL-83 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.0840	55	0.1205	44.2					1
	TCGA-KM-8 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.1323	65	0.0183	38.4	Event Type:	Loss		•	1
	TCGA-KM-8 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0763	88	0.0	31.1					1
	TCGA-KM-8 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0916	1	0.0	0.0	chr9:1-	-100000	00		1
	TCGA-KM-8 Proc	tessed Da	atabase Pr	Processed	Database Processing T	0.1023	31	0.0	36.0	0	100000	00		1
	TCGA-KM-8 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0636	148	0.0189	36.1					1
	TCGA-KM-8 Proc	cessed Da	atabase Pr	Processed	Database Processing T	0.0733	97	0.0	36.9					11
	TCGA-KM-8 Proc	essed Da	atabase Pr	Processed	Database Processing T	0.0997	l ol	0.0						1
Folgstod Same									I	.abel: Chr9	9p Loss			
Selected Samp	ies: 05 / 05									,				
Ready									I	Complete	elv coverer	4		



Cancel

### **Chr9p Concordance - Regions**

🔉 Concordan	ice								_	
Regions Circu	ular Plot 🗍 Geno	ome   Chromoso	me							
Export T	хт	view Annotations	s Modify vie	ew	Enrichment	Select				
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	i 0	88.235	NaN	
		chr9:8,330,	p24.1 - p23	CN Loss	1,158,499	88.235	i 0	88.235	NaN	
		chr9:10,309	p23	CN Loss	1,072,773	88.235	i 0	88.235	NaN	
		chr9:11,470	p23	CN Loss	235,705	88.235	i 0	88.235	NaN	
		chr9:12,041	p23 - p22.3	CN Loss	3,468,967	88.235	i 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		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	
	I	11.01.00	1949			00.050	"	00.050		
	Classic	P-Value T	hreshold 0.05	Differential T	hreshold 25.0	% Probe-level p-val	ues Calculate	Cancel		

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### Chr9p Concordance - Genome



**BioDiscovery** 

### Chr9p Concordance – Circular Plot

Na Concord	ance						Conce	ordanc	e							<u> </u>
Regions Cit	rcular Plot   Gen	ome Chromoso	me				Regions	; Circula	ar Plot   Genome	Chromosome						
Export	TXT	View Annotations	s Modify vi	ew	Enrichment	Select	3									
Display	Label	Region	Cytoband Location	Event 🛆	Region Le 🛆	Weighted Avg. Freq. i	Q Chr9	9p Loss								
		chr18:54,69	q21.31	CN Loss	450,560		A CHL	1,CNTN6	CNTN4-AS2,CN	TN4,IL5RA,TRNT	1,CRBN,LRRN1	SETMAR, SU	WF1,ITPR1-AS	S1,ITPR1,EGO	T,BHLHE40-A	S1,BHLHE40,ARI
		chr18:73,57	q23	CN Loss	519,538		B PRO	S1,ARL	13B,STX19,DHFR	L1,NSUN3,LINC	00879,MTHFD2F	21,MIR8060,EP	PHA6,ARL6,CI	RYBG3,MINA	GABRR3,OR	AC2,OR5H1,OR
		chr18:48,96	q21.2	CN Loss	541,543		C COL	_6A4P2,0	COL6A5,COL6A6	PIK3R4,ATP2C1	,ASTE1,NEK11	LOC339874,	VUDT16P1,NU	DT16,SNORA	58,MRPL3,CP	NE4,MIR5704,AC
		chr18:44,57	q21.1	CN Loss	719,376		D CT64	4,SI,SLIT	RK3,BCHE,ZBBX	SERPINI2,WDR4	49,PDCD10,SER	PINI1,LOC646	168,GOLIM4,E	EGFEM1P,MIR	551B,MECOM	TERC,ACTRT3,N
		chr19:34,29	q13.11	CN Loss	721,757		E PLEM	KHG4B,L	RRC14B,CCDC12	27,SDHA,LOC10	2467073,PDCD	6,AHRR,C5ort	f55,EXOC3,PP	7080,SLC9A	3,MIR4456,LO	C100996325,CEF
		chr7:145,95	. q35	CN Loss	727,380		F EMB	3,PARP8	LOC100287592,L	_OC642366,ISL1	,PELO,ITGA1,IT	GA2,MOCS2,	LOC257396,F	ST,NDUFS4,N	/IR581,ARL15	HSPB3,SNX18,L
		chr18:76,95	q23	CN Loss	738,913		G LOC	101928	569,PLK2,GAPT,L	_OC101928600,F	RAB3C,PDE4D,P	ART1,DEPDC	1B,ELOVL7,E	RCC8,NDUFA	F2,SMIM15,C1	C-436P18.1,ZSV
		chr7:145,06	q35	CN Loss	843,977		H DDX	(11L5,VV	ASH1,FAM138C,F	FOXD4,CBWD1,0	C9orf66,DOCK8	KANK1,DMR	11,DMR13,DM	RT2,SMARCA	42,FLJ35024,1	LDLR,KCNV2,K
		chr18:45,43	q21.1	CN Loss	1,134,999		I PIG	ER4P2-0		286297,AQP7P1,	,FAM27E3,FAM	Z/B,ANKRD20		COMPLEXES	UDN ACTIONS	2352,PGM5P2,Li
		chr7:153,13	q36.2	CN Loss	1,253,019		J SCG	20203P,	4D LIDI D D0414 A	ZNF599,LOC400	SAMENI SAM	ZNE3U, ZNE73	92,GRAMD1A 20001.2 MDID4	USCNIB, HPN,	110170 MID00	MIDI ETZC MIDI
		chr19:32,55	q13.11	CN Loss	1,304,754		L LOC	1020A	LINCOOLS8 MIR15	SSHC MIR155 LIN	JC00515 MRDI 3	0 IAM2 ATD4		D CVVR1 AD	00470,MIR33A	TSS MIRLET / C, WIRT
		chr19:43,54	q13.31	CN Loss	1,647,575		M ETS2	21.0010	1928398 AF0648	358 7 PSMG1 BR	1000313,MIRES	2 BR/MD1_AS	3, GADEA, AFI		86508 SH3BGE	C21orf88 B3G
		chr21:23,73	q21.1 - q21.2	CN Loss	1,854,860			2,20010	1020000,71 0040	, omor, or		2,014101-740	, , inclui, in		.0000,0110001	.,02101100,000,
		chr19:41,37	q13.2	CN Loss	1,917,039						~					
		chr21:32,74	q22.11	CN Loss	1,944,659					~ ×	1					
		chr21:34,72	q22.11 - q22.12	CN Loss	1,959,557					2		1 m				
		chr7:151,14	q36.1 - q36.2	CN Loss	1,991,814				ی کُر ک			- <b>(</b> )				
		chr18:74,76	. q23	CN Loss	2,060,209					N N						
		chr21:20,70	q21.1	CN Loss	3,004,515					J V						
		chr21:36,73	q22.12 - q22.2	CN Loss	3,444,159											
		chr7:146,69	q35 - q36.1	CN Loss	3,959,878				<u>و المع</u>			/B	- Co			
	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				<del>ا</del>							
	LOC339622,.	chr21:26,18	q21.2 - q22.11	CN Loss	6,535,973					R	5 A	///	4			
	EMB,PARP8,.	chr5:49,520	q11.1 - q11.2	CN Loss	7,809,323				14	E	<b>A</b> 38	F F				
	ETS2,LOC10.	chr21:40,19	q22.2 - q22.3	CN Loss	7,934,255					þ	۳ 🐙					
	COL6A4P2,	. chr3:129,86	q22.1 - q26.1	CN Loss	32,263,804				2	)	\ //					
	CT64,SI,SLI	. chr3:162,62	q26.1 - q29	CN Loss	35,396,146								5 5			
	PROS1,ARL.	. chr3:93,543	q11.1 - q22.1	CN Loss	36,220,273				2							
	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				42		G M		9			
	PTGER4P2-	cbr9:65-630	a12 - a34 3	CNLoss	75 582 497				-							
										01 <b>- 4</b>	8					
	Comt	pined 💌 P-	-Value Threshold	0.05 Differ	ential Threshold	25.0 %										

**BioDiscovery** 

# Comparing groups to find significant differences

# What changes are significant and unique to each group?



### Comparison – Metastasis (Clinical\_M)



### Comparison – M0 v MX

Nexus C	Copy Number	- NCI Training	) Session (Human NCBI Buil	137)	_ @ ×
ile Nexus	s DB Help	Evtornal Data Î	Populta [ Novus DR ]		
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Mud	Delete	VIGW	Decails	Comparison	
			Steps 1. Select Factors 2. Select Compariso	Select Comparison Baseline   Comparison Baseline   Comparison Baseline   Carcel     Organization     Details     Comparison Baseline     Carcel     Paired   Details     Comparison Baseline     Comparison Baseline     Comparison Baseline     Carcel     Paired   Details     Comparison Baseline     Paired     Details     Done     Cancel	
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				BioDiscovery © 2013	

### Comparison – M0 v MX - Regions

N clinical_M	- <mx> vs. <m0:< th=""><th>&gt;</th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th></m0:<></mx>	>									
Regions Gen	nome Chromosome										
Export	TXT View A	Annotations	Modify vie	w Enric	hment						
Region	Cytoband Location	Event	Region Length	Freq. in <mx>(%)</mx>	Freq. in <m0>(%)</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	<u> </u>
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	
i na		lenn.	4 000 400			50.000	••••		10	<u>-</u>	
	Classic	P-Value Threst	nold 0.05	Differential Thresho	ld 25.0 %	Probe-level p	values Calculate		Ca	ncel	

**BioDiscovery** 

### Comparison – M0 v MX - Genome



**BioDiscovery** 

### **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

scovery

### **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

over

- 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?



Reprove junction:         Verticity          Verticity         <	🎦 Invasion - <mi< th=""><th>(BC&gt; vs. <n< th=""><th>MIBC&gt;</th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th>_ 8</th></n<></th></mi<>	(BC> vs. <n< th=""><th>MIBC&gt;</th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th></th><th>_ 8</th></n<>	MIBC>											_ 8
Eges (n)         Ordan ducation         Fers (n, Region of Trans, netWork)         Pres, netWork(%)         Differion of hode-weigh-size         pice (n, Rick (%))         Gene Symbols         Garcerian           h11151 402,5551 504,505         0:2.2.02.3         Ordan 100,000         20.2.02.3.3         Ordan 100,000         20.2.001,000         0.000         Operating (%),000         0.000	Regions Genome	Chromosom	e											
Image: Cycloberd Lacokin, Even, Pagen Langh, Freq. n. 440EC (N)         Difference         Produit         Regin Langh, Freq. n. 440EC (N)         Difference         Freq. n. 440EC (N)         Difference (N)         Difference (N)         Difference (N)         Difference (N)         Difference (N)         Difference (N)         Di	Export TXT	View	Annotations	Modify v	riew I	Enrichment								
ch1137,32,559197,04500       q22       q23       Q1 Gan       q20,500       q27,500       No       q27,253       No       q27,253       No       q27,253       q23       q24       q23       q23       q24       q24       q23       q24       q23       q24       q24       q25       q34,83       No       q26       q1       q00       q24       q24 <td< th=""><th>Region</th><th></th><th></th><th>Event</th><th>Region Length</th><th>Ereq. in <math>\langle MIBC \rangle (\%)</math></th><th>Freq. in <nmibc>(%)</nmibc></th><th>Difference</th><th>Probe-level p-value</th><th>n-value</th><th>a-bound</th><th>% of CNV Overlap</th><th>Gene Symbols</th><th>CancerGeneCensu</th></td<>	Region			Event	Region Length	Ereq. in $\langle MIBC \rangle (\%)$	Freq. in <nmibc>(%)</nmibc>	Difference	Probe-level p-value	n-value	a-bound	% of CNV Overlap	Gene Symbols	CancerGeneCensu
ch1159 67132-169 015802       cp2 - q2.3       ONGA       289,580       17.647       95.5	chr1:157.432.553-15	57.504.503	g23.2	CN Gain	71,950	17,647	45	-27,353	NaN	1.42E-02	1.0	0 CADN	M3, LOC100131825, DARC	
ch1139.1158.519.12.112       ch139.1158.519.12.112       ch139.1158.519.12.114       ch139.12.804       ch139.1158.519.12.114       ch139.12.804       ch139.112.804       ch1189.112.804       ch1180.111.802       ch189.111.802       ch189.111.	chr1:158.627.312-15	58,915,892	a23.2 - a23.3	CN Gain	288,580	17.647	52.5	-34.853	NaN	3.28E-03	1.0	0 VANG	5L2, SLAMF6, CD84, SLAMF1, CD4	
bit 150 000 \$268 - 000 \$262         cols         cols         cols         cols         cols         cols         cols         cols         cols           ch1161 728 \$25 - 613 \$37,71         ccl 3         cl 46 an         17,477         22.5         -34.85         HMI         2786-40         1.0         0         -           ch1161 728 \$25 - 613 \$37,71         ccl 3         cl 46 an         179,477         22.5         -34.85         HMI         2786-40         1.0         0         -         AMT         AMT <td>chr1:159,911,585-15</td> <td>59,912,192</td> <td>q23.3</td> <td>CN Gain</td> <td>607</td> <td>26.471</td> <td>55</td> <td>-28,529</td> <td>NaN</td> <td>1.82E-02</td> <td>1.0</td> <td>100 FCGR</td> <td>12B</td> <td>FCGR2B</td>	chr1:159,911,585-15	59,912,192	q23.3	CN Gain	607	26.471	55	-28,529	NaN	1.82E-02	1.0	100 FCGR	12B	FCGR2B
h1111272285101       23.3       OVGN       90.982       17.447       95.8       94.83       NM       2.266.03       1.0       0         h11141555251614343731       624.1       O1 Gan       6,537       20.888       47.5       26.612       NM       2.765.02       1.0       0       PAM788         h111415555251643254764       p21.31       O1 Gan       6,537       20.888       47.5       26.612       NM       2.765.02       1.0       0       PAM788       VRIS	chr1:160,003,628-16	60,016,522	q23.3	CN Gain	12,894	20.588	52.5	-31.912	NaN	7.71E-03	1.0	100 ATF6	ì	
ch:114158,255-149,384,31       Q4.1       Q4 Gain       179,476       20.588       Q7.5       -26.512       NNI       2.708-02       1.0       0       PAM788         ch:1146,505,999-164,513,335       Q4.1       Q4.0       G.533       Q2.58       Q7.5       26.512       NNI       2.708-02       1.0       0       PAM788         ch:136,525,979-164,513,335       Q2.131       CNLoss       1.593,183       35.244       10       25.247       NNI       3.365:03       7.116-01       0.698/DOC3, MARF, RMISB, VFRBP, RADA, RAPT, PBM189, VFRBP, RADA	chr1:161,732,835-16	61,823,417	q23.3	CN Gain	90,582	17.647	52.5	-34.853	NaN	3.28E-03	1.0	0		
ch:114/13/95/96/97-14/51/358         cP4.1         Ch Gan         6.337         20.588         47.5         26.912         Nell 2.076-02         1.0         0            ch:346.516.015-86.625-543         p21.31         Chicos         1.0528         35.39         7.5         27.79         NeN         3.656-07         7.11E-01         0.0569         0.0569         DCFR5, UAL2, MIRTI, COTAL, UQC           ch:345.017.297-58.259.79         p21.1         Chicos         558,678         55.294         10         25.294         NeN         1.11E-02         7.11E-01         0.0569         DCFR5, UAL2, MIRTI, COTAL, UQC           ch:355.07.297-58.259.79         p21.1         Chicos         558,678         35.234         10         25.257         NeN         1.486-02         7.11E-01         0.0496/CTR8, SEU, CACNA203           ch:355.07.267.01         P1.778         27.16         1.00         P1T         P1T         P1T           ch:355.07.267.01         P1.176         15         26.176         NeN         1.756-02         7.11E-01         0.0499/MIR.290         P1T         P1T           ch:355.07.267.01         P1.176         15         26.176         NeN         1.756-02         7.11E-01         0.0359         NMIR 29.07.11E-01         0.0	chr1:164,155,255-16	64,334,731	q24.1	CN Gain	179,476	20.588	47.5	-26.912	NaN	2.70E-02	1.0	0 FAM7	78B	
ch:34:813.015-64,265.93         p21.31         ONLoss         109,820         35:29         7.5         27.79         NeN         3.85E-03         7.11E-01         ODE/NEE         OPERFE         UCUL, NIE71, CUTAL, UCC           ch:35:307.247.255.77         p21.1         ONLoss         358,675         35:294         10         25:294         NeN         1.11E-02         7.11E-01         0.0488 DCCS, MANF, RBM158, PREBP, RADCA         RADCA           ch:35:307.247.255.77         p21.1         ONLoss         358,675         36:23         12.5         25.735         NeN         1.43E-02         7.11E-01         0.048 ACTR3, SELV, CANADOS           ch:35:307.247.247         Ouery dimonsational sectors         1.325,553         38:23         12.5         25.735         NeN         1.43E-02         7.11E-01         0.048 ACTR3, SELV, CANADOS           ch:35:307.247.247         Ouery dimonsational sectors         95,957         41.176         15         26.176         NeN         1.75E-02         7.11E-01         0.038 Phy MISP3, TUBEL, FAM2296, LAMA4,           ch:119.99.026.231         ONLoss         5.339         441.176         15         26.176         NeN         1.75E-02         7.11E-01         0.038 Phy MISP3, TUBEL, FAM2296, LAMA4,           ch:119.99.026.247         ONLoss	chr1:164,506,999-16	64,513,336	q24.1	CN Gain	6,337	20.588	47.5	-26.912	NaN	2.70E-02	1.0	0		
ch:35.01/2.452.245.010       p21.31 - p21.1       CNLos       1,334,183       35.294       10       25.244       NAM       1115-02       7.11F-01       0.069 DCG3, MAMP, REMISE, VPEPP, RADS4 BAP1, PBRM1         ch:35.07/2.454.253.070       p21.1       CNLos       353,676       35.294       100       25.294       NAM       1.11E-02       7.11E-01       0.049 DCG3, MAMP, REMISE, VPEPP, RADS4 BAP1, PBRM1         ch:35.07/2.454.253.070       p21.1       CNLos       353,676       35.294       10.2       25.735       NAM       1.456-02       7.11E-01       0.049 ACTR6, SELK, CACM203         ch:35.07/2.454.253.070       Query all processed samples in protein       95,957       41.176       15       26.176       NAM       1.75E-02       7.11E-01       0.035 PM, WTS9, TUBE1, FAM229B, LAM44         ch:15.247.998.012.03.939       q22.11       QUery all processed samples in protein       55,339       44.115       15       26.176       NAM       1.75E-02       7.11E-01       0.235 PM, WTS9, TUBE1, FAM229B, LAM44         ch:15.247.998.013.937.939       q22.11       QUies       56.557       41.1176       115       26.176       NAM       1.75E-02       7.11E-01       0.235 PM, WTS9, TUBE1, FAM229B, LAM44         ch:15.247.947.945.939.049       q2.51.07       QUies       56.6570 <td>chr3:48,518,015-48,</td> <td>,626,543</td> <td>p21.31</td> <td>CN Loss</td> <td>108,528</td> <td>35.294</td> <td>7.5</td> <td>27.794</td> <td>NaN</td> <td>3.85E-03</td> <td>7.11E-01</td> <td>OPFKF</td> <td>B4, UCN2, MIR711, COL7A1, UQC</td> <td></td>	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	OPFKF	B4, UCN2, MIR711, COL7A1, UQC	
ch:35:37:24-54:25:90       p21.1       CNLoss       388,675       35.294       10       25.735       NAM       1.11E-01       0.045/CR8, SELV, CACNAD3       PHT         ch:35:70:274-56       Show Regio       711E-01       2.043/M1154, RC22111, M18939, RC21       PHT         ch:35:70:274-56       Show Regio       274,095       38.235       12.5       25.735       NaM       1.458-02       7.11E-01       6.171 PHT       PHT         ch:35:70:274-074       Query diprocessed samples in protein       95,957       41.176       15       26.176       NaM       1.75E-02       7.11E-01       0.04FMC2       PHT         ch:112,122,07       Query diprocessed samples in protein       95,957       41.176       15       26.176       NaM       1.75E-02       7.11E-01       0.04FMC2       PAMC2         ch:112,122,07       Query diprocessed samples in protein       2,216,091       41.176       15       26.176       NaM       1.75E-02       7.11E-01       0.04FMC2       PAMC2       PAMC2         ch:115,941,942,952,120,034,346       q22.1       Q22.1       Q22.2       Q230,381       41.176       15       26.176       NaM       1.75E-02       7.11E-01       0.35E/PM W193, UBER1, FAM2296, LAMA4,         ch:115,941,741,7454,7496	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	0.698DOCK	K3, MANF, RBM15B, VPRBP, RAD54	BAP1, PBRM1
dr3:59,76642.28       0.201       r.1.42       1,325,553       39.255       12.5       25.755       New 1,43502       7.11E-01       2.983/N1764, EPC2-T1, MIR3938, EPC2,         dr3:59,765,659       Show Region       274,095       38.225       12.5       25.735       New 1,43502       7.11E-01       6.171 FHIT       FHIT         chei:10,212,67       Query diabuse       274,095       38.235       12.5       25.735       New 1,43502       7.11E-01       100 FHIT       FHIT         chei:10,212,07       Query diabuse       41.176       15       26.176       New 1,75502       7.11E-01       0.48Mx22       CMEX         chei:11,527,794+117,551,815       q2.216,091       41.176       15       26.176       New 1,75502       7.11E-01       0.48Mx22       CMEX         chei:11,527,794+117,551,815       q2.21.0       QLvs       x55,570       41.176       15       26.176       New 1,75502       7.11E-01       0.48Mx24          chei:115,227,994+117,551,815       q2.2.1       CNLoss       556,570       41.176       15       26.176       New 1,75502       7.11E-01       0.282FM2A, FBX030, LOC1005757, SFPR         chei:15,261,715,125,350       q24.2       CNLoss       3,698,570       41.176       15	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 ACTR	R8, SELK, CACNA2D3	
chr:sp:99,699,699         Show Region         22,009         38.235         12.5         25.735         NaN         1.436-02         7.11E-01         6.171 [HTT         HTT           chr:sp:02,12,00         Query all processed samples in protect         147,800         41.176         15.26,176         NaN         1.755-02         7.11E-01         0.00PHT         HTT         HTT           chr:sp:12,12,07         Query all processed samples in protect         2,216,091         41.176         15         26.176         NaN         1.755-02         7.11E-01         0.04RMC2         chr:sp:12,02,07         0.048MC2         1.000 [HTT         HTT         HTT           chr:sp:12,02,07,080         242.1 - q2.2.2         NL0s         2,230,831         41.176         15         26.176         NaN         1.755-02         7.11E-01         0.0353 PM, WIS93,70E-01,175V         1.000 [HT         1.000 [HT <td>chr3:55,107,267-56</td> <td>432,820</td> <td>n14-3</td> <td>CNLoss</td> <td>1,325,553</td> <td>38.235</td> <td>12.5</td> <td>25.735</td> <td>NaN</td> <td>1.43E-02</td> <td>7.11E-01</td> <td>2.983 WNT</td> <td>5A, ERC2-IT1, MIR3938, ERC2,</td> <td></td>	chr3:55,107,267-56	432,820	n14-3	CNLoss	1,325,553	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	2.983 WNT	5A, ERC2-IT1, MIR3938, ERC2,	
Chr360.094.718         Copy         User         147,880         41.176         115         26.176         NNN         1.75E-02         7.11E-01         100 PHT         PHT           Chr6110.32.07.7         Query diabase         95,957         41.176         15         26.176         NNN         1.75E-02         7.11E-01         0.04RMC2         0.04	chr3:59,786,589-	Show Regio	n	- • F	274,095	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	6.171 FHIT		FHIT
checitog212,67         Query all processed samples in processed sampresind processed samples in procesed samples in processe	chr3:60,064,718-	Сору			147,880	41.176	15	26.176	NaN	1.75E-02	7.11E-01	100 FHIT		FHIT
chef:12,32,07       Query data/set       2,216,091       41.176       15       26.176       NaN       1.75E-02       7.11E-01       0.363 <pvn, fam2296,="" lama4,<="" td="" tubei,="" wisp3,="">         chef:115,247,964-117,551,815       q22.1       Q2Lo       NLos       5,23,334       44.116       115       26.618       NaN       1.75E-02       7.11E-01       0       0         chef:145,025,460-145,602,030       q24.2       CN Los       566,570       44.1176       115       26.616       NaN       1.75E-02       7.11E-01       0       0       0         chef:145,025,460-145,602,030       q24.2       CN Los       566,570       41.176       115       26.176       NaN       1.75E-02       7.11E-01       0</pvn,>	chr6:109,212,675	Query all pr	rocessed samples in pr	roject	95,957	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0 ARM(	C2	
chr6115,247,99-117,551,815       q22.1 - q22.2       CNLoss       2,303,831       41,176       15       26.176       NaN       1.75E-02       7.11E-01       0       0         chr6119,962,052-120,039,366       q22.1 - q22.2       CNLoss       52,334       441.18       17.5       26.618       NaN       2.07E-02       7.11E-01       0       0       0         chr6119,962,052-120,039,366       q24.2       CNLoss       566,570       41.176       15       26.176       NaN       1.75E-02       7.11E-01       0.0       UTRN         chr61150,911,437-151,190,460       q24.2       CNLoss       3,698,570       41.176       15       25.735       NaN       1.45E-02       7.11E-01       0.5788       5R1         chr61152,611,147-152,449,675       q25.1       CNLoss       500,892       41.176       15       26.176       NaN       1.75E-02       7.11E-01       5.758       5R1	chr6:112,132,07:	Query data	base		2,216,091	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.363 FYN,	WISP3, TUBE1, FAM229B, LAMA4,	
chr6119982,052-120,034,38       q2.31       NL 0s       \$52,334       44.18       17.5       26.618       NAN       2.07E-02       7.1E-01       0       UTRN       Image: Constraint of the constrai	chr6:115,247,984-11	17,551,815	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	
chr6:145,035,460-145,002,003       q24.2       NLoss       S566,570       41.176       155       26.176       NAN       1.75E-02       7.11E-01       OLITEN       Alternation of the state	chr6:119,982,052-12	20,034,386	q22.31	CN Loss	52,334	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	0		
chr6:145,636,936-149,335,506       q24.2 - q25.1       CN Loss       3,698,570       41.176       15       26.176       NAM       1.75E-02       7.11E-01       0.352       PM2A, FBX030, LOC100507557, SHPR         chr6:150,811,437-151,190,400       q25.1       CN Loss       379,023       38.235       1.25       25.75       NAM       1.43E-02       7.11E-01       0       PLEKHG1         chr6:153,124,340-153,625,232       q25.2       CN Loss       168,528       41.176       155       26.176       NAM       1.75E-02       7.11E-01       0.71E-0X       0.74E-0X       0.74E	chr6:145,035,460-14	45,602,030	q24.2	CN Loss	566,570	41.176	15	26.176	NaN	1.75E-02	7.11E-01	OUTRN	J	
chr6:150,811,437-151,190,460       q25.1       CN Loss       379,023       38.235       1.2.5       25.735       NAN       1.43E-02       7.11E-01       0 <plekhg1< th="">       1.2.5         chr6:152,281,147-152,2449,675       q25.1       CN Loss       168,528       41.176       1.5       26.176       NAN       1.75E-02       7.11E-01       0.5788       ESR1       0         chr6:155,261,147-162,449,075       q25.2       CN Loss       500,092       41.176       1.5       26.176       NAN       1.75E-02       7.11E-01       0.2.7457       TIAM2, CDN20, TFB1M, NO3,       0         chr6:155,1621-162,445,1528       q26.2       Q5.2       Q5.2       Q5.00       41.176       1.55       26.161       NAN       1.75E-02       7.11E-01       0.2.044 GPA14, PARX       0         chr6:165,1021-162,4451,528       q26       Q1.00       2.6.16       NAN       1.75E-02       7.11E-01       0.2.044 GPA14, PARX       0.0.0         chr6:166,737,766-167,071,567       q27       NLos       3.98,001       44.118       0.7.5       26.618       NAN       1.75E-02       7.11E-01       0.6.837       Corf118, PDE10A, LINC00473, LINC00</plekhg1<>	chr6:145,636,936-14	49,335,506	q24.2 - q25.1	CN Loss	3,698,570	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.352 EPM2	A, FBXO30, LOC100507557, SHPR	
chr6:152,281,147-152,449,675       q25.1       Q1Loss       168,52       41.176       Ch1       26.176       NAN       1.75E-02       7.11E-01       5.758       ENT       ENT       ENT         chr6:155,124,340-153,625,232       q25.2       Q1Loss       500,892       41.176       G1B       26.176       NAN       1.75E-02       7.11E-01       0.211       EXOS, MTRF1L, RG517       Q       Q         chr6:155,142,142,430,552,323       q25.2       Q25.3       Q1Loss       30,0502       41.176       Q1D       26.176       NAN       1.75E-02       7.11E-01       Q2.7457       TIAM2, CLDN20, TFB1M, NOX3,       Q       Q         chr6:165,473,589-163,371,49       q26       Q1Loss       896,992       41.176       Q1D       26.616       NAN       1.75E-02       7.11E-01       Q.044       APAR2, PACRG       Q       <	chr6:150,811,437-15	51,190,460	q25.1	CN Loss	379,023	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	OPLEK	HG1	
chr6:153,124,340-153,625,232       q25.2       QN Los       500,892       41.176       Chr       26.176       NaN       1.75E-02       7.11E-01       D.211       EXX5, MTRF1L, RG517       Additional state         chr6:155,167,354-157,120,56       q25.2 - q25.3       QN Los       1.653,222       41.176       Chr       26.176       NaN       1.75E-02       7.11E-01       C.71E-01       C.7	chr6:152,281,147-15	52,449,675	q25.1	CN Loss	168,528	41.176	15	26.176	NaN	1.75E-02	7.11E-01	5.758ESR1		
chr6:155,467,354-157,120,566       q25.2 - q25.3       CNL os       1,653,22       41.176       CM       26.176       NAM       1.75E-02       7.11E-01       27.475       TAM2, CLDN20, TFB1M, NOX3,       A         chr6:161,551,021-162,451,528       q26       CNL os       900,507       44.118       CM       26.618       NAM       2.07E-02       7.11E-01       2.044 A/GPAT4, PARK2       A         chr6:162,475,389-163,371,481       q26       CNL os       896,092       44.176       CM       26.618       NAM       1.75E-02       7.11E-01       2.044 A/GPAT4, PARK2       ARK2, PACRG       A         chr6:166,247,661653,362       q27       CNL os       1.911,666       41.176       CM       26.618       NAM       1.75E-02       7.11E-01       G.0637       G.06118, PDE10A, LINC00473, LINC00473, LINC0047, LINC00473,	chr6:153,124,340-15	53,625,232	q25.2	CN Loss	500,892	41.176	15	26.176	NaN	1.75E-02	7.11E-01	0.211 FBXO	95, MTRF1L, RGS17	
chr6:161,551,021-162,451,528       q26       NL oss       9900,507       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       NL oss       896,092       44.118       17.5       26.618       NAN       1.75E-02       7.11E-01       4.0582 PARK2, PACKG       4.016         chr6:164,640,702-166,552,686       q27       NL oss       1.911,666       41.176       26.176       NAN       2.07E-02       7.11E-01       4.0582 PARK2, PACKG       4.016         chr6:166,734,766-167,071,567       q27       NL oss       1.911,666       44.118       17.5       26.176       NAN       2.07E-02       7.11E-01       4.0582 PARK2, PACKG       4.016         chr6:166,734,766-167,071,567       q27       NL oss       3.36,801       44.118       17.5       26.176       NAN       2.07E-02       7.11E-01       0.08F56KA2-1T1, MIR1913, RP56KA2       0.016       0.0	chr6:155,467,354-15	57,120,566	q25.2 - q25.3	CN Loss	1,653,212	41.176	15	26.176	NaN	1.75E-02	7.11E-01	27.457 TIAM	12, CLDN20, TFB1M, NOX3,	
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       ARK2, PACRG       ARK2, 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       Corf118, PDE10A, LINC00473, LINC00       Ark2, PACRG       Ark2, PACRG, PACRG, PACRG, PACRG, PACRG, PACR	chr6:161,551,021-16	62,451,528	q26	CN Loss	900,507	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	2.044 AGPA	AT4, PARK2	
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       Coorf118, 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.RP56KA2-1T1, MIR1913, RP56KA2         chr6:166,233,620-168,290,566       q27       CN Loss       57,236       47.059       17.55       29,559       NaN       1.09E-02       7.11E-01       0.0       RP56KA2-1T1, MIR1913, RP56KA2         chr10:13,789,296-13,874,667       p13       CN Loss       57,236       47.059       17.55       29,559       NaN       1.09E-02       7.11E-01       0.054       RMD4A       0.056         chr10:13,789,296-13,874,667       p13       CN Loss       40,056       38.235       12.55       CA11       1.99E-02       1.0       0.54       RMD4A       0.0       0.0       RMD4A       0.0 <th< td=""><td>chr6:162,475,389-16</td><td>63,371,481</td><td>q26</td><td>CN Loss</td><td>896,092</td><td>41.176</td><td>15</td><td>26.176</td><td>NaN</td><td>1.75E-02</td><td>7.11E-01</td><td>40.582 PARK</td><td>2, PACRG</td><td></td></th<>	chr6:162,475,389-16	63,371,481	q26	CN Loss	896,092	41.176	15	26.176	NaN	1.75E-02	7.11E-01	40.582 PARK	2, PACRG	
chr6:166,734,766-167,071,567       q27       QN Loss       336,801       44.118       17.5       26.618       NaN       2.07E-Q       7.11E-01       QRP56KA2-TT1, MIR1913, RP56KA2         chr6:168,233,620-168,290,856       q27       QN Loss       57,236       47.059       17.5       29,559       NaN       1.09E-Q       7.11E-01       100            chr10:13,789,296-13,874,667       p13       QN Gai       85,371       41.176       12.5       26.176       NaN       1.75E-Q       1.0       0.65F RMD4A          chr13:34,934,183-34,974,239       q13.3       QN Gai       31,207       Q       38.235       12.5       25.735       NaN       1.45E-Q       7.11E-01       0.05F RMD4A       MH11       MVH11         chr16:15,824,760-15,855,967       p13.11       QN Gai       31,207       Q       27.55       NaN       1.45E-Q       1.0       MEA, MAB2111, MIR548F5       MVH11         chr16:15,973,870-15,992,796       p13.11       QN Gai       31,926       Q       27.55       NaN       6.2EE-Q       1.0       MMEA, MAB2111, MIR548F5       MVH11         chr16:15,973,870-15,992,796       p13.11       QN Gai       31.892       QC1       MVH11       MVH11      <	chr6:164,640,702-16	66,552,368	q27	CN Loss	1,911,666	41.176	15	26.176	NaN	1.75E-02	7.11E-01	6.387 C6orf	f118, PDE10A, LINC00473, LINC00	
chr6:168,233,620-168,290,856       q27       QN Loss       57,236       47.059       17.5       29.559       NaN       1.09E-02       7.11E-01       100	chr6:166,734,766-16	67,071,567	q27	CN Loss	336,801	44.118	17.5	26.618	NaN	2.07E-02	7.11E-01	0RPS6	KA2-IT1, MIR1913, RP56KA2	
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       1.45E-02       7.11E-01       NEEA, MAB21L1, MIR548F5         chr16:15,824,760-15,855,967       p13.11       CN Gain       31,207       O       25.735       NaN       1.36E-03       1.0       MPH11       MYH11         chr16:15,973,870-15,992,796       p13.11       CN Gain       18,926       O       27.75       NaN       6.2EE-04       1.0       MECA, MAB21L1, MIR548F5       MH11         chr16:15,973,870-15,992,796       p13.11       CN Gain       18,926       O       27.55       NaN       6.2EE-04       1.0       MBEC1       MECA	chr6:168,233,620-16	68,290,856	q27	CN Loss	57,236	47.059	17.5	29.559	NaN	1.09E-02	7.11E-01	100		
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 MBEA, 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       4	chr10:13,789,296-13	3,874,667	p13	CN Gain	85,371	41.176	15	26.176	NaN	1.75E-02	1.0	0.54 FRME	D4A	
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       100         Image: 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       100	chr13:34,934,183-34	4,974,239	q13.3	CN Loss	40,056	38.235	12.5	25.735	NaN	1.43E-02	7.11E-01	0 NBEA	4, MAB21L1, MIR548F5	
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           I <t< td=""><td>chr16:15,824,760-15</td><td>5,855,967</td><td>p13.11</td><td>CN Gain</td><td>31,207</td><td>0</td><td>25</td><td>-25</td><td>NaN</td><td>1.36E-03</td><td>1.0</td><td>ОМУН1</td><td>11</td><td>MYH11</td></t<>	chr16:15,824,760-15	5,855,967	p13.11	CN Gain	31,207	0	25	-25	NaN	1.36E-03	1.0	ОМУН1	11	MYH11
	chr16:15,973,870-15	5,992,796	p13.11	CN Gain	18,926	0	27.5	-27.5	NaN	6.22E-04	1.0	100 ABCC	1	
	4													
Peaks Only T P-Value Threshold 0.05 Differential Threshold 25.0 % Prohe-level n-values Calculate Concel			De la companya	eaks Only	P-Value Th	reshold 0.05 Diff	erential Threshold 25	0 %	Probe-level n-values	Calculate		Cancel		
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Projects   Samples   All Samp	0814923-52749106						1.28% (686 of 53396 ) sam
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Di Otuvetuduetti OOO		Project Name	Regions	Length	Classification	Notes	
	TCGA-14-1454-01A-01D-051	TCGA-GBM-Hudson	chr3:46,	6,564,268			
2e-01wstwkzhi_003	TCGA-14-1459-01A-01D-051	TCGA-GBM-Hudson	chr3:49,	3,856,187			
2e-01wstwkzhl_005	ICGA-19-1392-01A-01D-051	ICGA-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			
	TCGA-02-0113-01A-01D	TCGA-GBM-Stanford	chr3:30,	25,692,837			
	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			
p24.3	3 p2	1.31 p14.2 p14	<b>i.1</b> p12.3				q23 q24 q28.1 q28 q2
Query:chr3:50,814,923-	-52,749,106		Results range:chr	3:0-199,298,3	72		View:chr3:0-199,298,372

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### What if I have my own data?

## **Project Set-up and Processing**



### Launch Nexus Copy Number



### **Creating A New Project**

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

🗏 Create Ne	w Project	×
Project Name	Bootcamp BLCA Project	
Location	C:\Users\Andrea J. O'Hara\Desktop	
Organism	Human	
Build	NCBI Build 36.1	
	Create Cancel	

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• Click 'Create'

**Discovery** 

### Load Data

M	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 Sample	Quality Manually Altered	Ę
Load Data		^
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### Parameters Affected by the Load Data Guide

🕅 Add Sample Data 🛛 🗙	
Copy number Seq. Variation	
Select data type Affymetrix MIP	
Set processing settings based on the following and process	
<ul> <li>For mosaic samples such as cancer (possible increased false-positive rate)</li> <li>For non-mosaic samples</li> </ul>	
Stringency: I I Lenient Average Stringent	
Perform Systematic Correction	
Array Types: Catalog Affy MIP 330K 20110301 Catalog Affy MIP2 333K 20121226 Select Files Catalog Affy MIP2 20110512	
Samples	
Done Cancel	
Ready	
BioDiscovery	•

- 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	×
Copy number Seq. Variation	
Select data type Affymetrix MIP	
Set processing settings based on the following and process	Look in: 📙 Nexus Bootcamp Data 💌 🎓 🖽 📰
<ul> <li>For mosaic samples such as cancer (possible increased false-positive)</li> <li>For non-mosaic samples</li> </ul>	Recent Items
Stringency: I I I Lenient Average Stringent	Desktop
Perform Systematic Correction     Array Types: Catalog Affy MIP2 20110512	My Documents
Select Files Remove	Computer
Janpios	File name:         Training_Copy_Number.txt         Open           Network         Files of type:           Cancel
Done Cancel	
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### **Pre-processing of Data in Nexus**

Systematic correction

Before

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4.50 2.25 -4.25 2.00 4.00 1.75 3.75 1.50 3.50 1.25 3.25 1.00 3.00 .75 2.75 2.50 2.75 -50 1.50 -75 125 -1.00 1.00 -1.25 .75 -1.50 .50 -1.75 25--2.00 --2.25 --25 -2.50 5 6 7 8 9 10 11 12 13 14 15 16 17 18 19 20 21 22 1

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After

### **Process Samples**

🧏 Nexus Copy Number - Bootcamp BLCA Pr	oject (Human NCBI Build 36.1)			
File Nexus DB Help				
Data Set Comparisons External Data Nexus D	B]			
Load  Select View	Delete Reset Duplicate F	actors  Query Tools		
	Sample	Status	Data Type	Quality
<b>N</b>	G5M1083178	Processed	Affymetrix MIP	1.069 🔺
	G5M1083186	Processed	Affymetrix MIP	0.925
<b>v</b>	G5M1083189	Processed	Affymetrix MIP	0.554
	G5M1083191	Processed	Affymetrix MIP	0.686
	G5M1083194	Processed	Affymetrix MIP	0.996
	G5M1083200	Processed	Affymetrix MIP	1.075
	G5M1083212	Unprocessed	Affymetrix MIP	
<b>N</b>	G5M1083217	Unprocessed	Affymetrix MIP	
	G5M1083227	Unprocessed	Affymetrix MIP	
	G5M1083230	Unprocessed	Affymetrix MIP	
	G5M1083235	Unprocessed	Affymetrix MIP	
<b>N</b>	G5M1083238	Unprocessed	Affymetrix MIP	
	G5M1083242	Unprocessed	Affymetrix MIP	
	G5M1083243	Unprocessed	Affymetrix MIP	
	G5M1083249	Unprocessed	Affymetrix MIP	
Selected Samples: 15 / 15				
Loading probes for GSM1083212				Cancel

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### **Import Sequence Variants Data**

NX	Add Sample Data	×			
Copy numb	er Seq. Variation				
Select data	type Affy MIP Somatic Mutation	~		í.	
✓ Auto	omatic Affy MIP Somatic Mutation Affymetrix OncoScan MBZIP				
Selec	t File MAF VCF Represented Mutations				
Samples					
	Done Cancel				
Ready			1	1 1	
ery					
	BioDiscovery © 20	13			

### **Import Sequence Variants Data**

Select         Yiew         Delte         Reset         Duplicate         Factors         Query         Tools           V         Sample         Status         Data Type         Seq. Variation Status         Seq. Variation Data Type         Quality           V         GSM1083189         Processed         Affymetrix MIP         Processed         MAF         0.055           V         GSM1083189         Processed         Affymetrix MIP         Processed         MAF         0.055           V         GSM1083194         Processed         Affymetrix MIP         Processed         MAF         0.056           V         GSM1083194         Processed         Affymetrix MIP         Processed         MAF         0.056           V         GSM1083191         Processed         Affymetrix MIP         Processed         MAF         0.066           V         GSM108320         Processed         Affymetrix MIP         Processed         MAF         0.047           V         GSM1083227         Processed         Affymetrix MIP         Processed         MAF         0.047           V         GSM1083221         Processed         Affymetrix MIP         Processed         MAF         0.049           V         GSM108323		nal Data   Results   Nexus DB					
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Image: CSM1083166ProcessedAffymetrix MIPProcessedMAF0.928Image: CSM1083194ProcessedAffymetrix MIPProcessedMAF0.939Image: CSM1083178ProcessedAffymetrix MIPProcessedMAF0.0686Image: CSM1083191ProcessedAffymetrix MIPProcessedMAF0.0686Image: CSM1083200ProcessedAffymetrix MIPProcessedMAF0.0686Image: CSM1083200ProcessedAffymetrix MIPProcessedMAF0.0677Image: CSM1083200ProcessedAffymetrix MIPProcessedMAF0.0477Image: CSM1083200ProcessedAffymetrix MIPProcessedMAF0.0477Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0479Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0479Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0479Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0479Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0479Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0477Image: CSM1083212ProcessedAffymetrix MIPProcessedMAF0.0477Image: CSM1083217ProcessedAffymetrix MIPProcessedMAF0.0477Image: CSM1083217ProcessedAffymetrix MIPProcessedMAF <td< td=""><td></td><td>GSM1083189</td><td>Processed</td><td>Affymetri× MIP</td><td>Processed</td><td>MAF</td><td>0.554</td></td<>		GSM1083189	Processed	Affymetri× MIP	Processed	MAF	0.554
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Image: Mage: M		G5M1083178	Processed	Affymetrix MIP	Processed	MAF	1.069
Image: Constraint of Constra	<b>v</b>	G5M1083191	Processed	Affymetrix MIP	Processed	MAF	0.686
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Image: Constraint of Constra		G5M1083227	Processed	Affymetrix MIP	Processed	MAF	0.491
Image: Second		G5M1083212	Processed	Affymetrix MIP	Processed	MAF	0.449
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GSM1083235 Processed Affymetrix MIP Processed MAF 1.707		G5M1083217	Processed	Affymetrix MIP	Processed	MAF	0.408
	✓	GSM1083235	Processed	Affymetrix MIP	Processed	MAF	1.707
	V	GDM1083235	Processed		Processed	MAF	1.707





### 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

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• Can be entered manually or loaded from a tabdelimited text file in the Data Set tab

### Load Factors – Example Factors File

- 4	А	В	С	D	E	F	G	Н
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	Та	nephroureterectomy
4	GSM1083189	renal pelvis	FFPE urothelial carcinoma	78	male	2	Та	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	cystoprostatectom y
8	GSM1083212	bladder	FFPE urothelial carcinoma	79	male	3	T4a	cystectomy
9	GSM1083217	renal pelvis	FFPE urothelial carcinoma	75	female	2	ТЗ	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

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

White Papers:

**BioDiscovery** 

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

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