

Profiling Individual Patients in cBio Cancer Genomics Portal

Jianjiong Gao

Memorial Sloan-Kettering Cancer Center



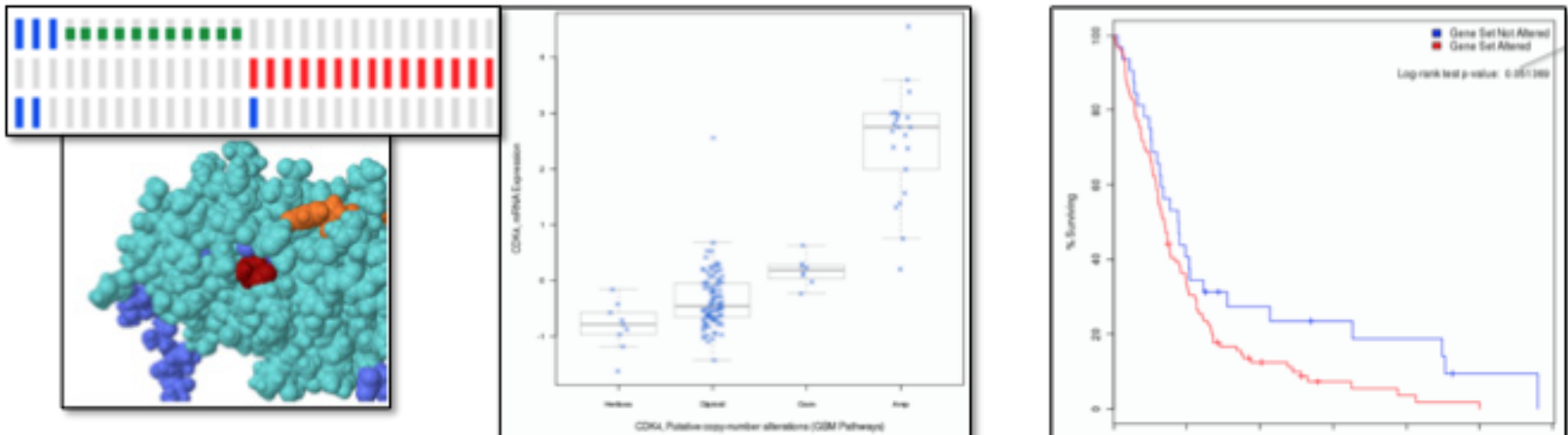
Memorial Sloan-Kettering
Cancer Center



cBio Cancer Genomics Portal *Enabling research with convenient access to data and analyses*

- **Remove barriers of access to cancer genomic data and analysis**
- **Make Cancer Genomics data available to a wide audience**
 - “Wet lab” biologists involved in functional studies
 - Clinical researchers
 - Computational biologists who want programmatic access to TCGA data
- **Facilitate explorative data analysis**
 - Quickly view genomic alterations and their patterns
 - Change parameters and iterate as easily as possible
 - Download of “data slices”
- **Key abstraction: “Altered gene”**

cbioportal.org



Patient-centric View: Motivation

Tumor samples tend to have hundreds or thousands of alterations

Which of these are relevant?

Can we use these relevant events to define the choice of treatment ultimately?

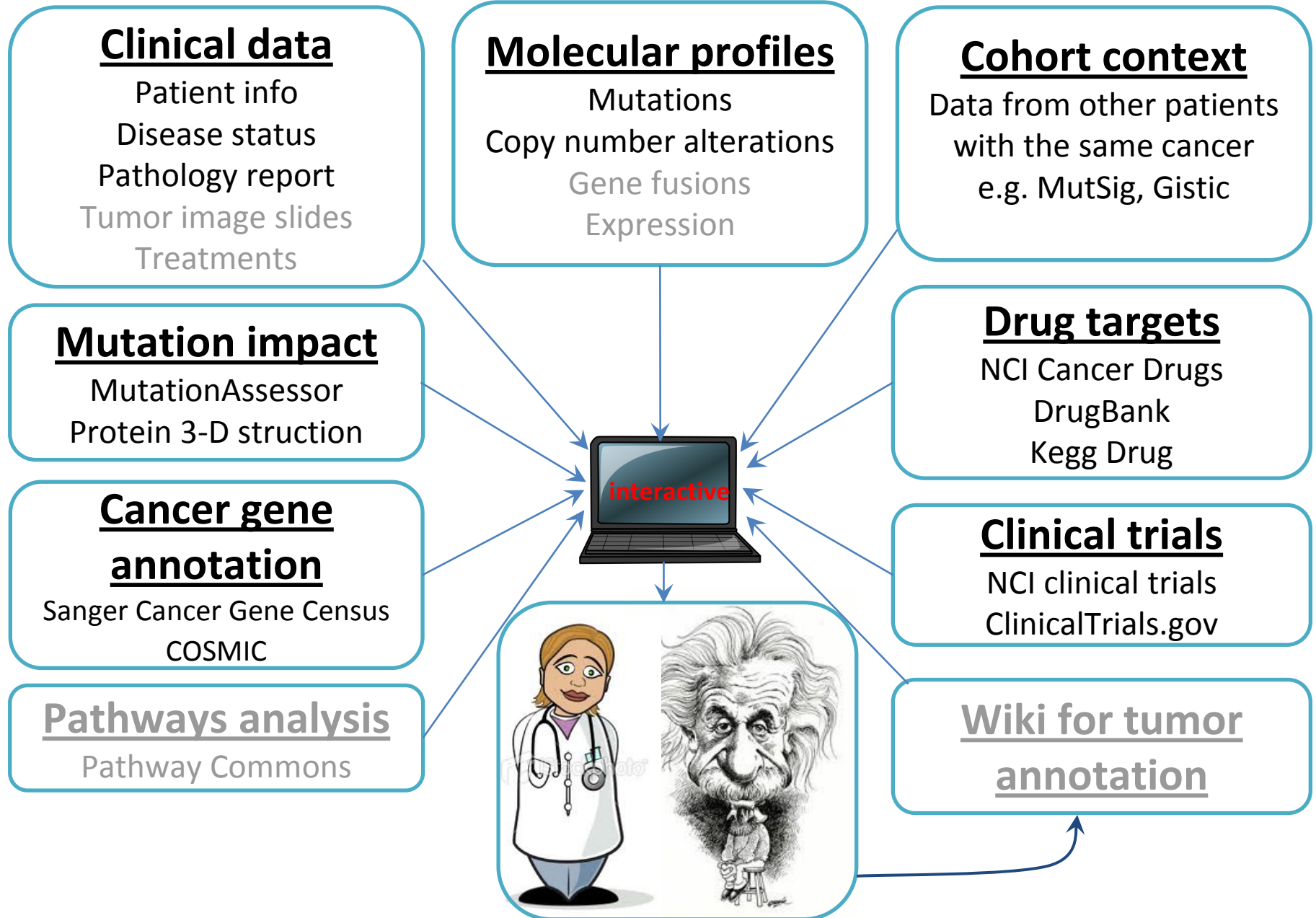
Experts are good at intuitively / manually identifying them

Can we **automate** parts of this process?

Can we **improve** this process?

Goal: Interactive system to facilitate personalized cancer research and ultimately guide patient treatment

Profiling an individual patient in cBio Portal



Example: TCGA Endometrial Cancer

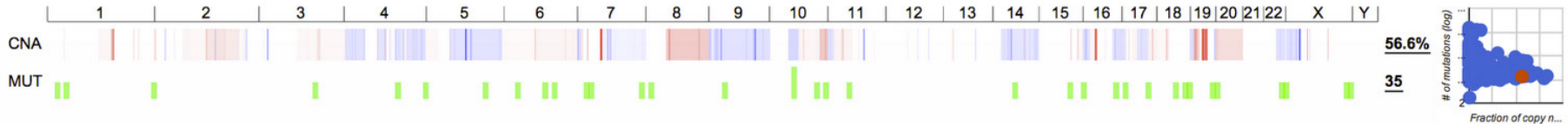
Case TCGA-BK-A0CC

TCGA-BK-A0CC FEMALE, 69 years old
Uterine Corpus Endometrioid Carcinoma (TCGA, Provisional), Serous, Stage III, Grade 3

[More about this patient](#)

Living (10 months), DiseaseFree (10 months)

Summary Mutations Copy Number Alterations Pathology Report



Mutations of interest (4 of 35)

Gene	Protein Change	Type	Cohort	COSMIC	FIS	3D
TP53	Q331*	NS		16		
PPP2R1A	S256F	MS		11	H	3D
FAT1	E314*	NS				
EPHA7	H408Q	MS			M	3D

Show all 35 mutations

Show 25 per page

CNA of interest (17 of 385)

Gene	CNA	% in Cohort
MCL1	AMP	
PSMD4	AMP	
ZNF687	AMP	
PI4KB	AMP	
RFX5	AMP	
PIP5K1A	AMP	
CCNE1	AMP	
BCL7C	AMP	
CTF1	AMP	
FBXL19-AS1	AMP	
FBXL19	AMP	
ORAI3	AMP	
SETD1A	AMP	
HSD3B7	AMP	
STX1B	AMP	
CEBPA	AMP	
EGFR	AMP	

Show all 385 CNAs

Show 25 per page

Example: TCGA Endometrial Cancer

Patient info & Clinical data

TCGA-BK-A0CC FEMALE, 69 years old More about this patient
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Example: TCGA Endometrial Cancer

More clinical data

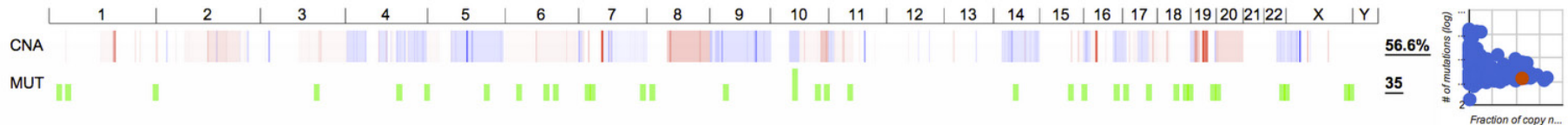
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2009stagegroup	Stage III
Age	69.0
data_core_sample	Y
data_gistic	Y
data_maf	Y
data_rna_seq	Y
Disease-free survival months	10
Disease-free survival status	DiseaseFree
histology	Serous
methylation_cluster	3
mhl1_hypermethylated	0
micro_rna_cluster	4
micro_rna_score	0.886489497
mrna_expression_cluster	1
msi_status_5_marker_call	MSS
msi_status_7_marker_call	MSS
mutation_rate_cluster	1_LOW
Overall survival months	10
Overall survival status	Living
tumor_grade	Grade 3

Example: TCGA Endometrial Cancer

Case TCGA-BK-A0CC

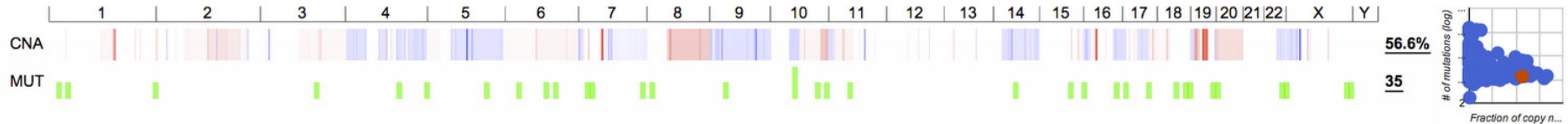
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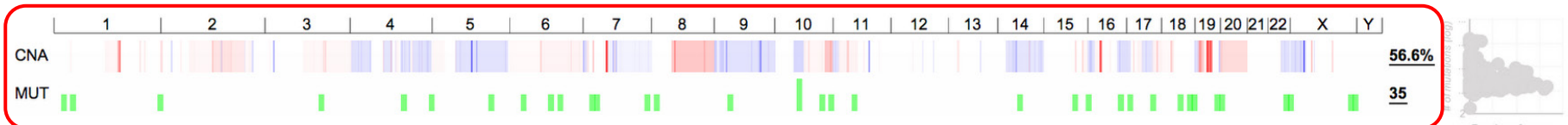
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Example: TCGA Endometrial Cancer

Genomic overview

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BCL7C	AMP	█
CTF1	AMP	█
FBXL19-AS1	AMP	█
FBXL19	AMP	█
ORAI3	AMP	█
SETD1A	AMP	█
HSD3B7	AMP	█
STX1B	AMP	█
CEBPA	AMP	█
EGFR	AMP	█

Show all 385 CNAs

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Example: TCGA Endometrial Cancer

Scatter plot of mutations vs CNA

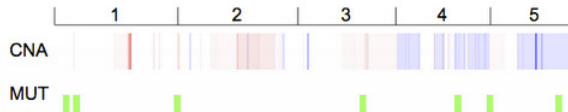
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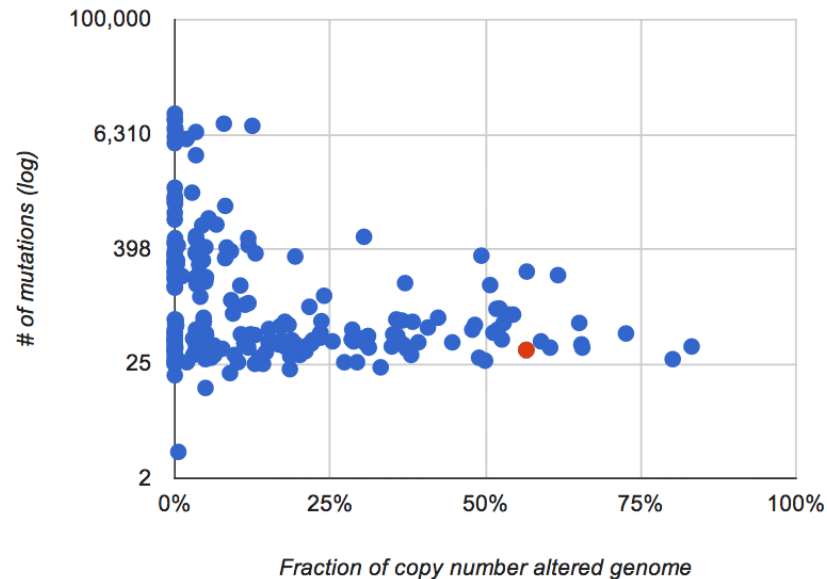


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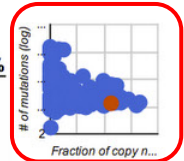
Mutation Count vs Copy Number Alterations



H-Axis scale: linear log

V-Axis scale: linear log

Each dot represents a tumor sample in [Uterine Corpus Endometrioid Carcinoma \(TCGA, Provisional\)](#).



Navigation and search controls for the mutation list, including a search bar and a list of mutation entries with circular icons.

per page

Example: TCGA Endometrial Cancer

Mutations of interest

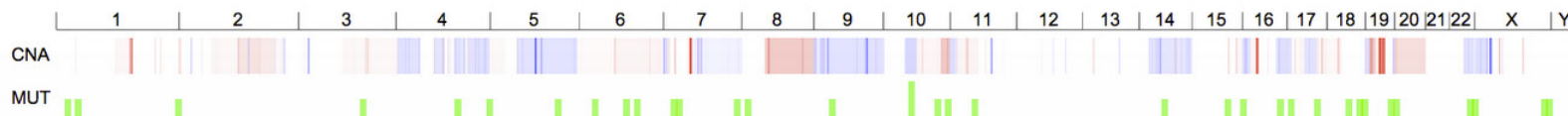
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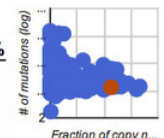
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Summary Mutations Copy Number Alterations Pathology Report



56.6%

35



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Example: TCGA Endometrial Cancer

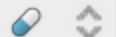






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










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Show per page  

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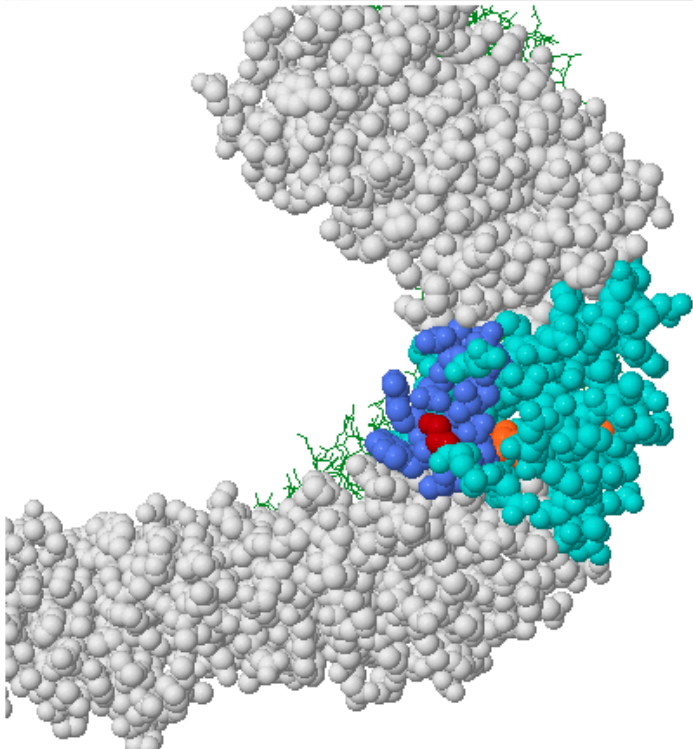
Example: TCGA Endometrial Cancer

Mutations of interest

TCGA-BK-A0CC FEMALE 69 years old
Uterine Corpus Endo

2AAA_HUMAN/197-282 : VEACVNIAQLLPQEDLEALVMPTLRQAAEDKSWRVRYMVADKFTELQKAVGPEITKTD
midline : VEACVNIAQLLPQEDLEALVMPTLRQAAEDKSWRVRYMVADKFTELQKAVGPEITKTD
1b3u:A/196-281 : VEACVNIAQLLPQEDLEALVMPTLRQAAEDKSWRVRYMVADKFTELQKAVGPEITKTD

specificity ■
conserved ■
neutral ■
unmapped ■
hetero ■
variant ■



Jmol

Summary Mut

Mutat

Gene

TP53

PPP2R1A

FAT1

EPHA2

Show all

More about this patient
10 months), DiseaseFree (10 months)

3D

3D

essor
equence Alignment

EGFR AMP

Show all 385 CNAs

Show 25 per page

Example: TCGA Endometrial Cancer

Copy number alteration of interest

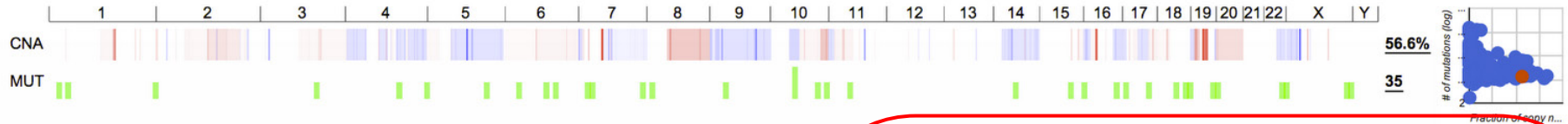
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Example: TCGA Endometrial Cancer

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TCGA-BK-A0CC FEMAL
Uterine Corpus Endomet

Summary Mutatio

CNA
MUT

Mutations of in

Gene Protei
Chan

TP53 Q331
PPP2R1A S256
FAT1 E314
EPHA7 H408

Show all 35 mutatio

More about this patient
, DiseaseFree (10 months)

of mutations (log)
56.6%
35
Fraction of copy n...

Q-value: 1.9e-18
Number of genes in the peak: 1

Show all 385 CNAs

Show 25 per page

Gene	CNA	% in Cohort	
MCL1	AMP	■	
PSMD4	AMP	■	⊗
ZNF687	AMP	■	⊗
PI4KB	AMP	■	⊗
RFX5	AMP	■	⊗
PIP5K1A	AMP	■	⊗
CCNE1	AMP	■	⊗
BCL7C	AMP	■	
CTF1	AMP	■	
FBXL19-AS1	AMP	■	
FBXL19	AMP	■	⊗
ORAI3	AMP	■	⊗
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HSD3B7	AMP	■	⊗
STX1B	AMP	■	⊗
CEBPA	AMP	■	
EGFR	AMP	■	⊗

Example: TCGA Endometrial Cancer

Copy number alteration of interest

Drug name: Cetuximab
Target: EGFR, FCGR3B, C1R, C1QA, C1QB, C1QC, FCGR3A, C1S, FCGR1A, FCGR2A, FCGR2B, FCGR2C, EGFR
Synonyms: IMC-C225; Erbitux; Cetuximab; Anti EGFR
FDA approved? Yes
Description: Epidermal growth factor receptor binding FAB. Cetuximab is composed of the Fv (variable; antigen-binding) regions of the 225 murine EGFr monoclonal antibody specific for the N-terminal portion of human EGFr with human IgG1 heavy and kappa light chain constant (framework) regions.
Data sources: NCI, PharmGKB, DrugBank
Clinical Trials: 204 clinical trials

Drug name: Trastuzumab
Target: ERBB2, EGFR, C1R, C1QA, C1QB, C1QC, C1S, FCGR1A, FCGR2A, FCGR2B, FCGR2C, FCGR3B, FCGR3A, EGFR
Synonyms: Trastuzumab; Anti HER2; Herceptin; Ig gamma-1 chain C region
FDA approved? Yes
Description: A recombinant IgG1 kappa, humanized monoclonal antibody that selectively binds with high affinity in a cell-based assay ($K_d = 5$ nM) to the extracellular domain of the human epidermal growth factor receptor protein. Produced in CHO cell culture.*
Data sources: NCI, PharmGKB, DrugBank
Clinical Trials: 146 clinical trials

Drug name: Gefitinib
Target: EGFR, EGFR
Synonyms: Iressa; Irressat; Gefitinib
FDA approved? Yes
Description: Gefitinib (originally coded ZD1839) is a drug used in the treatment of certain types of cancer. Acting in a similar manner to erlotinib (marketed as Tarceva), gefitinib selectively targets the mutant proteins in malignant cells. It is marketed by AstraZeneca under the trade name Iressa. [Wikipedia]
Data sources: NCI, PharmGKB, DrugBank, KEGG Drug
Clinical Trials: 36 clinical trials

EGFR

AMP

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25 per page

Example: TCGA Endometrial Cancer

Pathology report

TCGA-BK-AOCC FEMALE, 69 years old

Uterine Corpus Endometrioid Carcinoma (TCGA, Provisional), Serous, Stage III, Grade 3

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Living (10 months), DiseaseFree (10 months)

Summary

Mutations

Copy Number Alterations

Pathology Report

UID:642AAC78-9889-4044-8F8D-94132576C2D
TCGA-BK-AOCC-01A-PR Redacted



SPECIMEN

- A. Uterus, cervix, bilateral tubes and ovaries, bilateral parametria, upper vagina
 - B. Additional left vaginal margins
 - C. Right pelvic lymph node
 - D. Right periaortic lymph nodes
 - E. Left pelvic lymph nodes
 - F. Left periaortic lymph nodes
 - G. Omentum
 - H. Left pericolic biopsy
 - I. Bladder peritoneum
 - J. Cul-de-sac peritoneum
 - K. Right pelvic biopsy
 - L. Left pelvic biopsy
 - M. Right pericolic biopsy
 - N. Left diaphragm biopsy
 - O. Right diaphragm biopsy
- 1 CD. 0-3
adenocarcinoma, papillary serous 8460/3
Site: endometrium C St. 1 Lu
4/3/0*

CLINICAL NOTES

PRE-OP DIAGNOSIS: Endometrial cancer.
HISTORY: Endometrial and cervical biopsies papillary serous adenocarcinoma.

FROZEN SECTION DIAGNOSIS

A - There is tumor present in both the endometrium and cervix, cannot be certain on frozen section if separate processes or part of same process. There is both invasion and areas of high-grade carcinoma. [* **]

GROSS DESCRIPTION

A. Received fresh for frozen section and tissue procurement labeled "uterus, cervix, bilateral tubes and ovaries, bilateral parametria, upper vagina" is a 100 gm., 4.7 x 4.0

GROSS DESCRIPTION

X 4.0 cm. symmetrical uterine corpus with attached 3 cm. cervix and bilateral adnexa. The serosa is smooth to scabrous tan pink-red. The pericervical soft tissue margin is inked blue. The 4.0 x 3.8 cm. tan white ectocervix surrounds a patulous 2.5 x 1.0 cm. os. A rubbery tan red polypoid lesion protrudes from the os. On opening the posterior cervix contains an irregular, 3.5 x 2.0 cm. rubbery tan pink-red lesion. On sectioning the lesion is glistening tan white and appears to measure up to 1.3 cm. in thickness. The lesion appears to extend to within 0.8 cm. of the inked peripheral margin of the cervix. A rim of apparent tan white vaginal mucosa circumferences the ectocervix. The triangular, 4.0 x 3.2 cm. uterine cavity contains tan white tumor predominating within the fundus both anteriorly and posteriorly. On sectioning, the lesion has a maximal thickness of 1.1 cm. and appears confined to the



Example: TCGA Endometrial Cancer

Link to cancer study view

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Uterine Corpus Endometrioid Carcinoma (TCGA, Provisional)

Serous, Stage III, Grade 3

[More about this patient](#)

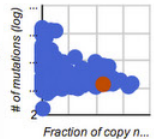
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Cancer Study View

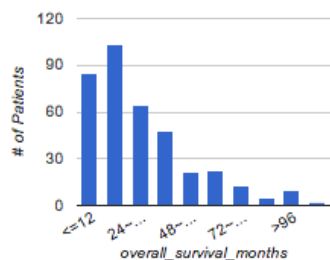
Interactive exploration of cohort data

Uterine Corpus Endometrioid Carcinoma (TCGA, Provisional) [Query this study](#)

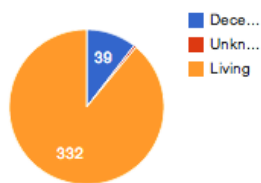
TCGA Uterine Corpus Endometrioid Carcinoma, containing 373 samples; raw data at the NCI.

Clinical Data Mutated Genes Copy Number Alterations

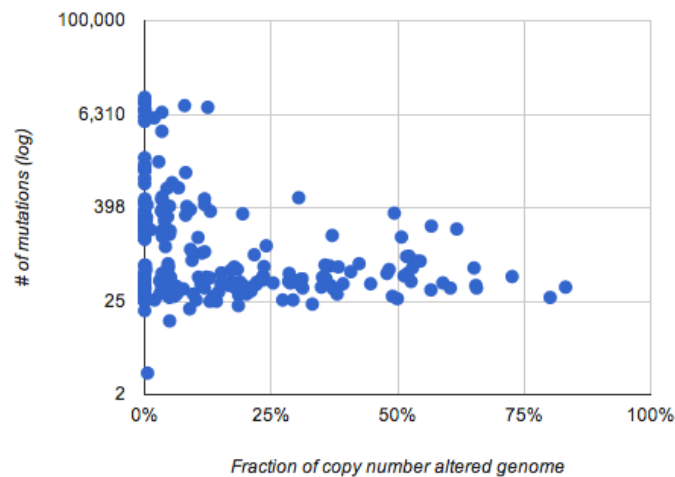
Overall survival months



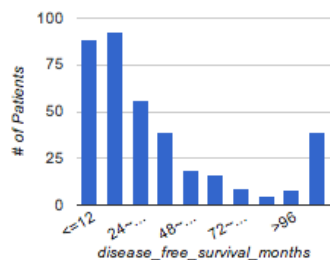
Overall survival status



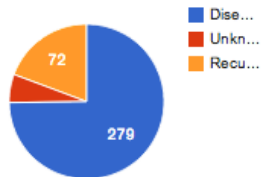
Mutation Count vs Copy Number Alterations



Disease free survival months

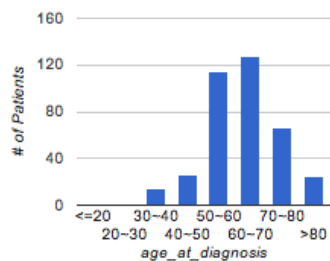


Disease free survival status

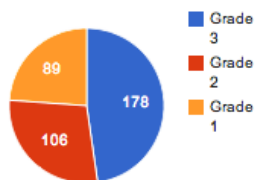


H-Axis scale: linear log
V-Axis scale: linear log

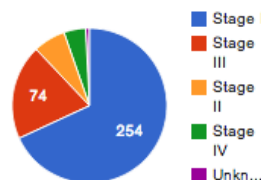
Age at diagnosis



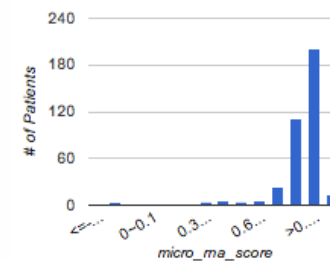
Tumor grade



2009stagegroup

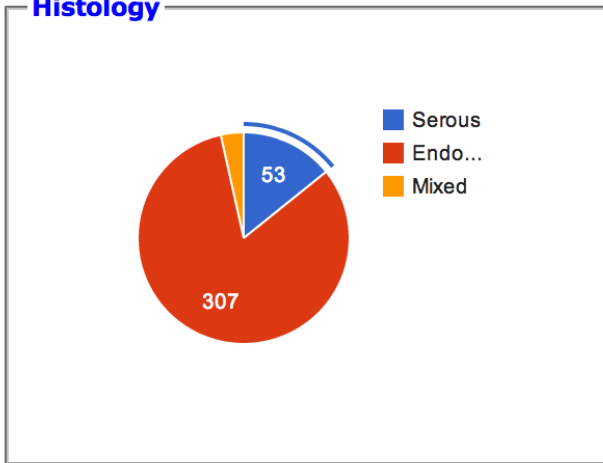


Micro rna score

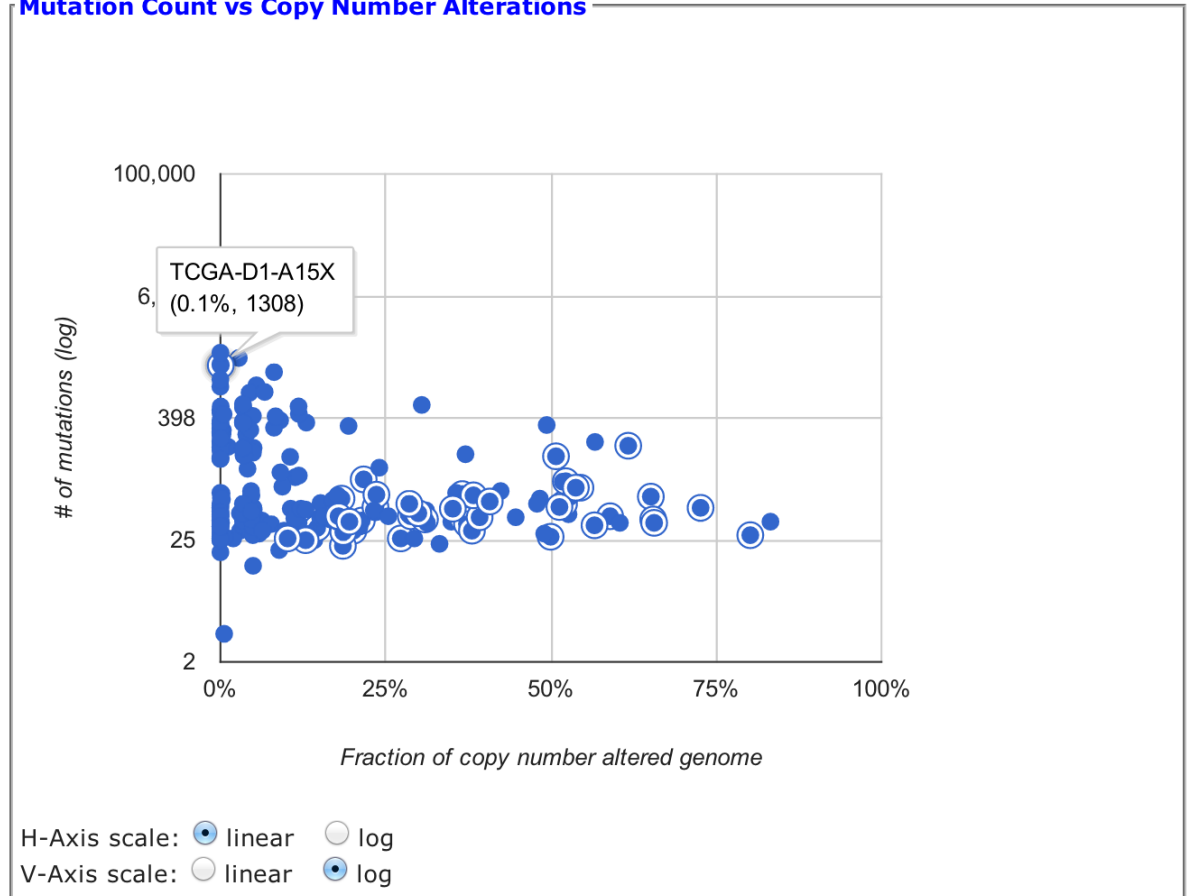


Plots are interconnected

Histology



Mutation Count vs Copy Number Alterations

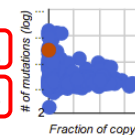
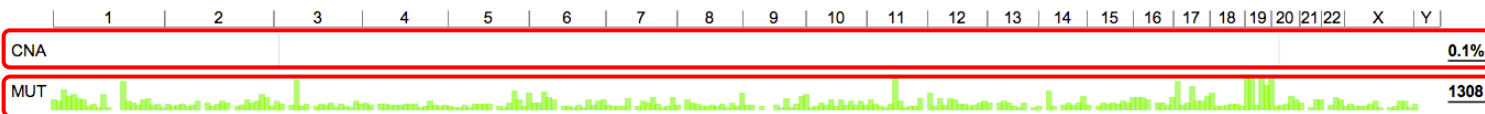


Summary

Mutations

Copy Number Alterations

Pathology Report



Mutations of interest (54 of 1308) ?

Search:

Gene	Protein Change	Type	Cohort	COSMIC	FIS	3D
KRAS	G13D	MS			2998	M 3D
FBXW7	R505C	MS			57	M 3D
PIK3CA	C420R	MS			26	M 3D
ARID1A	R2236P	MS			1	L
EPHA10	R150H	MS			1	L 3D
TSC1	D47G	MS			1	M
MLL2	L2113P	MS				N
MLL2	R1614W	MS				M
FGFR2	R664W	MS				M 3D
APC	N1122D	MS				L
ATM	M1644I	MS				L
MLL3	R4211Q	MS				L
MED12	V1588M	MS				M
TET1	H1999L	MS				M
SETD2	R2077Q	MS				L
SETD2	K1560R	MS				L 3D
CDH10	A184T	MS				M 3D
CREBBP	P1900A	MS				L
CREBBP	I1038M	MS				N
RASA1	R398*	NS				
RASA1	L680S	MS				M
SIN3A	L983P	MS				M
SIN3A	R485C	MS				M 3D
EPHA5	F637S	MS				L
GLI3	R366Q	MS				L

[Show all 1308 mutations](#)

Show 25 per page

CNA of interest (0 of 5) ?

Search:

Gene CNA % in Cohort

No CNA events of interest

[Show all 5 CNAs](#)

Show 25 per page

Cancer Study View

Significantly/Recurrently Mutated Genes

Clinical Data Mutated Genes Copy Number Alterations

144 significantly mutated genes by MutSig Search:

Gene	# Mutations	Q-value
PTEN	227	1.83e-12
PIK3CA	172	1.83e-12
ARID1A	94	1.83e-12
PIK3R1	100	1.83e-12
CTNNB1	80	1.83e-12
TP53	74	1.83e-12
KRAS	52	1.83e-12
CTCF	49	1.83e-12
FBXW7	46	1.83e-12
FGFR2	34	1.83e-12
SPOP	23	1.85e-12
PPP2R1A	30	8.29e-12
P2RY11	9	8.91e-9
CHD4	43	2.63e-7
CCND1	15	3.86e-7
FAM9A	20	1.280e-6
NFE2L2	15	6.270e-6
FOXA2	12	7.900e-6
ARID5B	34	8.690e-6
TNFAIP6	12	1.100e-5
MORC4	28	2.300e-5
CYLC1	27	2.800e-5
RBMX	14	7.300e-5
RASA1	30	1.200e-4
SOX17	7	4.600e-4

Show 25 per page (1 to 25 of 144)

Cancer Study View

Copy number alteration peaks

Clinical Data
Mutated Genes
Copy Number Alterations

80 copy number alteration peaks by **GISTIC2** Search:

AMP/DEL	Chr	Cytoband	# Genes	Genes	Q-value
DEL	19	19p13.3	70	GNA11 SH3GL1 AES DAPK3 EEF2 GNA15 GNG7 MATK GADD45B NFIC MAP2K2 SGTA TBXA2R THOP1 TLE2 S1PR4 APBA3 CHAF1A EBI3 HMG20B SEMA6B ZFR2 PIP5K1C TIMM13 SNORD37 TJP3 NMRK2 SLC39A3 ZBTB7A FZR1 SIRT6 PIAS4 LSM7 STAP2 CCDC94 C19ORF10 NCLN SPPL2B SHD ZNF77 CACTIN CELF5 FSD1 TLE6 ZNF556 UBXN6 DOHH CREB3L3 HDGFRP2 LMNB2 RAX2 MPND ATCAY ZNF554 MRPL54 LRG1 TMIGD2 TNFAIP8L1 ZNF57 MFSO12 GIPC3 DIRAS1 ZNF555 ANKRD24 C19ORF77 TMRSS9 CACTIN-AS1 PLIN5 PLIN4 C19ORF71	1.24e-32
AMP	8	8q24.21	1	MYC	1.15e-24
AMP	17	17q12	2	ERBB2 PGAP3	4.07e-23
AMP	3	3q26.2	1	MECOM	1.22e-19
AMP	19	19q12	1	CCNE1	1.89e-18
DEL	5	5q11.2	0		1.21e-17
DEL	1	1p36.11	9	ARID1A SFN NR0B2 NUDC GPN2 PIGV GPATCH3 ZDHHC18 C1ORF172	5.71e-17
DEL	4	4q34.3	1	LINC00290	3.36e-16
DEL	2	2q22.1	1	LRP1B	4.58e-16
AMP	3	3q26.2	4	MECOM TERC MYNN ACTRT3	1.01e-13
DEL	10	10q23.31	2	PTEN KLLN	6.05e-13
DEL	1	1p36.31	0		1.08e-11
DEL	6	6q26	1	PARK2	5.72e-11
DEL	16	16q23.1	1	WWOX	1.77e-1
AMP	1	1q21.3	5	PI4KB PSMD4 RFX5 PIP5K1A ZNF687	4.59e-1
DEL	17	17q11.2	4	NF1 EVI2A EVI2B OMG	1.51e-9
AMP	3	3p25.1	0		1.34e-8
AMP	20	20q11.21	4	PLAGL2 TM9SF4 POFUT1 TSPY26P	2.15e-8
AMP	10	10q22.2	7	KAT6B VDAC2 DUSP13 COMTD1 SAMD8 ZNF503-AS1 DUPD1	6.61e-8
AMP	1	1p22.3	1	DDAH1	1.01e-7
DEL	11	11p15.5	0		4.62e-7
AMP	12	12q13.2	1	ERBB3	2.502e-6

Short Demo

cBioPortal.org

Acknowledgements

Nikolaus Schultz

Benjamin E. Gross

Bülent Arman Aksoy

Selcuk Onur Sumer

Gideon Dresdner

Boris Reva

Ethan Cerami

Chris Sander

Douglas Levine

Workshop Session 7
Alexandria Room

Poster #65

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