



## IntOGen – Integrative Onco Genomics

<https://www.intogen.org/>

<http://bg.upf.edu/group/img/1-s2.0-S1535610815000574-main.pdf>

<http://www.nature.com/nmeth/journal/v10/n11/full/nmeth.2642.html>

# Home Page

 Search Downloads Analysis About Login

  
Integrative  
Onco  
Genomics

Search example | Show more examples

**Release 2014.12**

PlotTable

### IntOGen Mutations 2014.12

Cancer types and projects chart



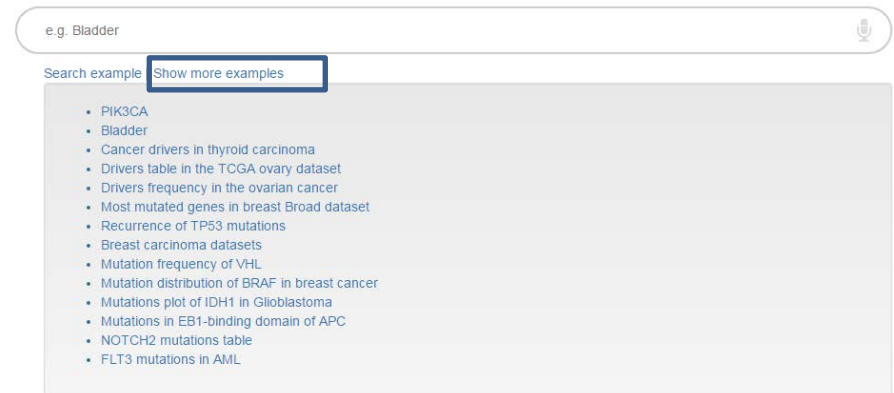
IntOGen collects and analyses somatic mutations in thousands of tumor genomes to identify cancer driver genes.

<b>Cancer Types</b>	28
<b>Projects</b>	48
<b>Samples</b>	6792
<b>Somatic mutations</b>	1341752
<b>Coding sequence mutations (CSMs) ⓘ</b>	
in driver genes	21648
in all genes	1341706
<b>Protein affecting mutations (PAMs) ⓘ</b>	
in driver genes	18649
in all genes	603770

Feedback & Support

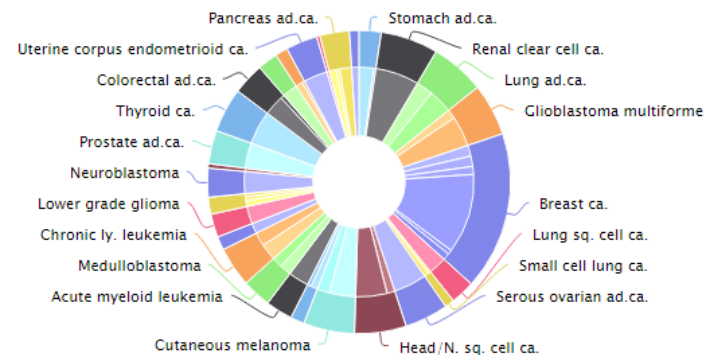
# Search Bar

- Examples provided for effective searches
- Home Page depicts cancer types with information in IntOGen
  - Pie chart or Table format
  - Can use this information to create effective search



IntOGen Mutations 2014.12

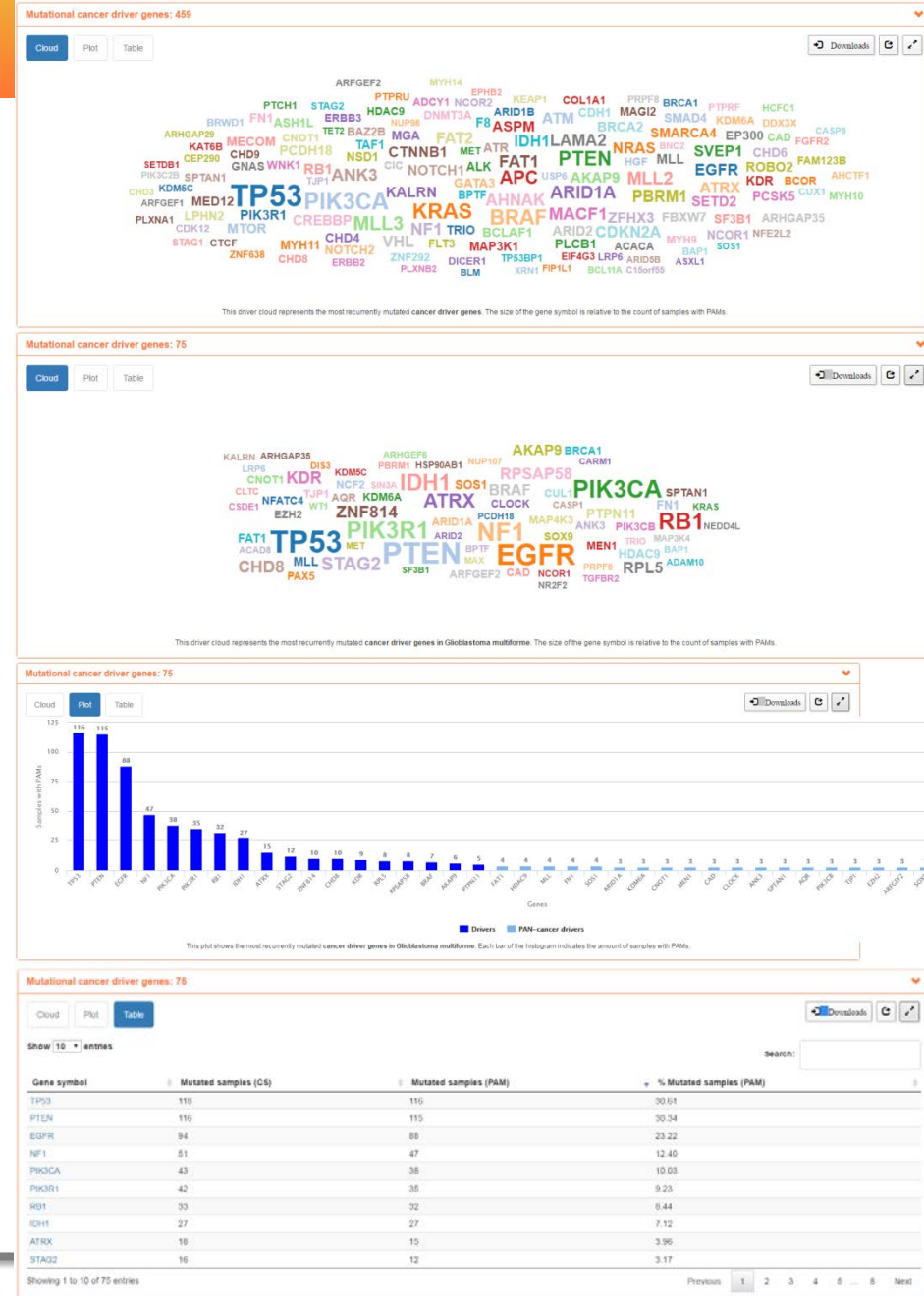
Cancer types and projects chart



# Mutational Cancer Drivers



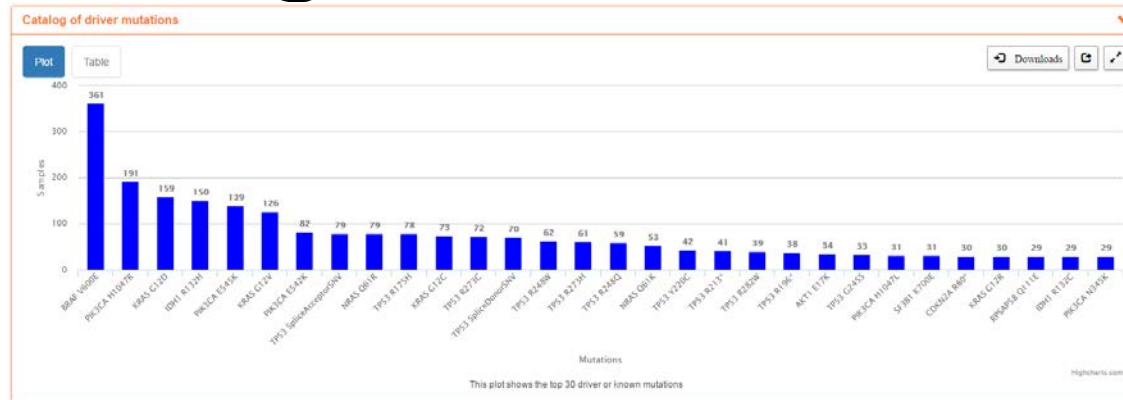
- Home page shows word cloud with the most frequently mutated cancer driver genes. (top right)
- Narrow search to see disease-specific cloud
  - Glioblastoma Multiforme (GBM) depicted on middle right
- Log in to download
  - Can sign up with Google Account
- Data is able to be saved
- View Data as Word Cloud, bar graph or table



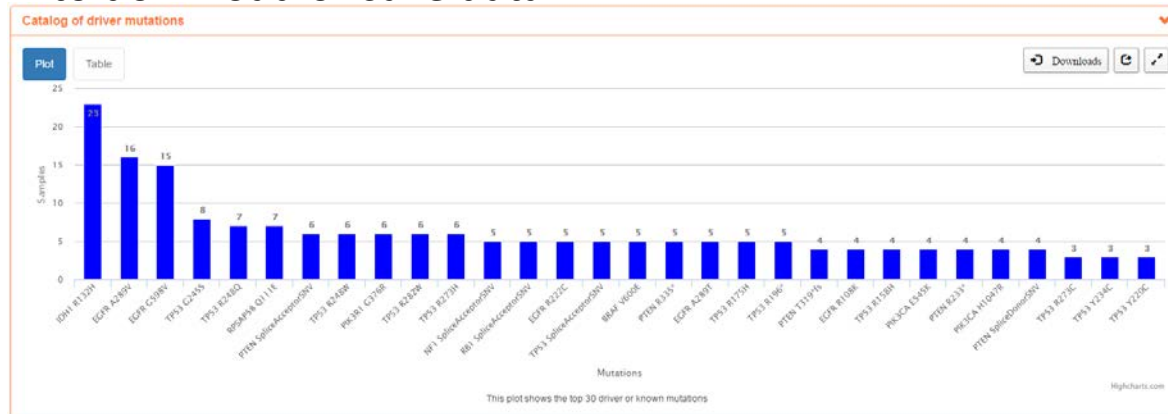
# Mutation Data and Driver Determination

- Tumor mutation data: from large projects (ICGC, TCGA, other publications).
- Driver Mutation Identification: Classification done in silico using a number of tools
  - OncodriveFM: <http://bg.upf.edu/group/projects/oncodrive-fm.php>
  - OncodriveCLUST: <http://bg.upf.edu/group/projects/oncodrive-clust.php>
  - MutSigCV: <http://archive.broadinstitute.org/cancer/cga/mutsig>
  - OncodriveROLE: <http://bg.upf.edu/oncodrive-role/>

# Catalog of Driver Mutations




- On Home Page (above), this table shows a Pan Cancer view of specific driver mutations.
- On Disease Specific page (below is table for GBM), this table shows driver mutations within your selected disease
  - Log in to download or save data.



# Projects Table

- On resulting page from search, this table is available
  - Links to project page (with information from that source) and links to project publications

Projects 				
Project	Authors	Source	Reference	Sample size
<a href="#">GBM_TCGA</a>	The Cancer Genome Atlas	TCGA SYNAPSE	<a href="#">doi:10.1038/nature07385</a>	290
<a href="#">GBM_JHU</a>	Johns Hopkins University	ICGC	<a href="#">PMID:18772396</a>	89

# Search for Gene

- Summary
- Gene Details
- Driver List
- Mutations per Cancer Type
- Mutations along protein sequence

**FLT3**

**FLT3** is detected as a mutational cancer driver in 1 cancer type: Acute myeloid leukemia and in the PAN-cancer analysis

**Reports**

- [Driver signals](#)
- [Mutation frequency](#)
- [Mutation distribution](#)

---

**Gene details**

FLT3

Ensembl id

ENSG00000122025

Mutated samples

Coding Sequence

163 (2.4%)

Protein Affecting

111 (1.6%)

Mode of action

Activating

Known driver

Yes



# Search for Gene

- Summary
- Gene Details
- Driver List
- Mutations per Cancer Type
  - Log in to download as .png or .csv
- Mutations along protein sequence
  - Can view different transcripts
  - Log in to download as .png or .csv



# Add Disease to Search Criteria

- Refines search criteria to only include mutations involved in cancer type specified.
  - Top right – all FLT3 mutations
  - Bottom right – FLT3 filtered for disease type

Acute myeloid leukemia

FLT3

FLT3 is detected as a mutational cancer driver in 1 Acute myeloid leukemia project

**Reports**

- Driver signals
- Mutation frequency
- Mutation distribution

**Cancer type details**

Acute myeloid leukemia

Projects	1
Samples	196
Coding sequence mutations (CSMs)	
in driver genes	409
in all genes	3330
Protein affecting mutations (PAMs)	
in driver genes	376
in all genes	1798

**Gene details**

FLT3

Ensembl id	ENSG00000122025
Mutated samples	
Coding Sequence	53 (27.0%)
Protein Affecting	28 (14.3%)
Mode of action	Activating
Known driver	Yes

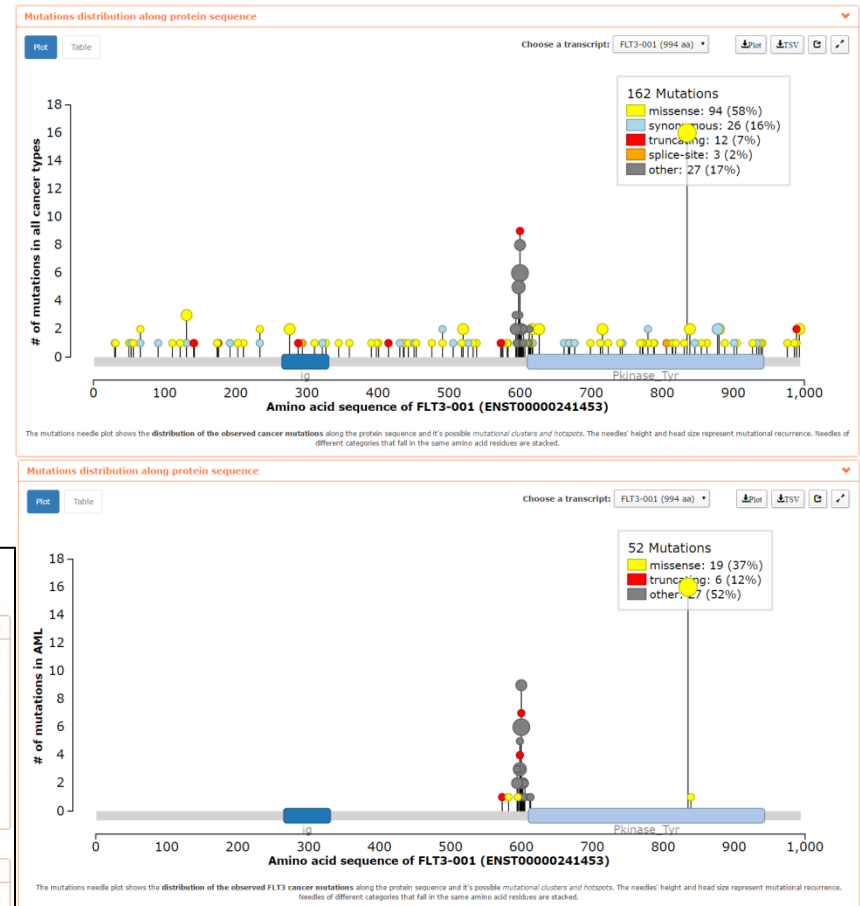
**Driver signals in 1 projects**

Show 10 entries

Project	Signals
Acute myeloid leukemia TCGA	<div>Rec</div> <div>FM</div> <div>Clust</div>

Showing 1 to 1 of 1 entries

Clust Clustered Mutations
 FM Functional Mutations
 Rec Recurrent Mutations

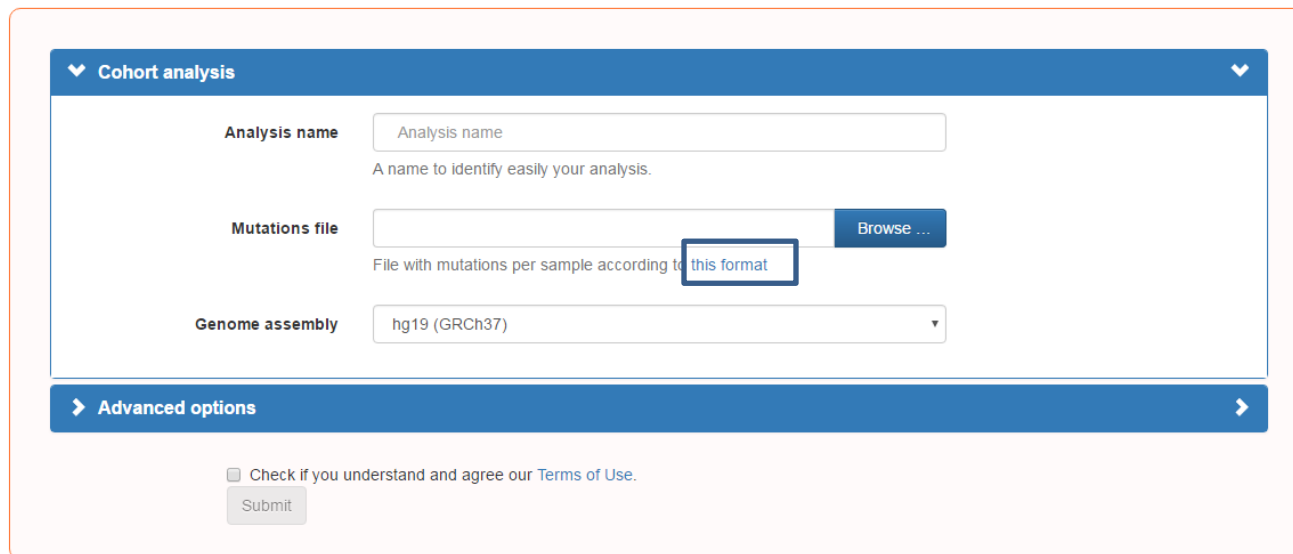


# Downloads Page

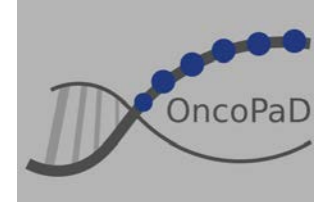
- Log into account to download databases
  - **Catalog of Driver Mutations (2016.5)**
    - Results of driver analysis using Cancer Genome Interpreter (See separate slide deck) and OncodriverMUT
    - Tamborero D., Rubio-Perez C., Deu-Pons J., Schroeder M., Vivancos A., Rovira A., Tusquets I., Albanell J., Rodon J., Tabernero J., Dienstmann R., Gonzalez-Perez A. and Lopez-Bigas N. **Cancer Genome Interpreter identifies driver and actionable alterations**. Manuscript in preparation
  - **Cancer Drivers Database (2014.12)**
    - In silico prescription of drugs based on somatic mutations
    - Rubio-Perez, C., Tamborero, D., Schroeder, MP., Antolín, AA., Deu-Pons, J., Perez-Llamas, C., Mestres, J., Gonzalez-Perez, A., Lopez-Bigas, N. [In silico prescription of anticancer drugs to cohorts of 28 tumor types reveals novel targeting opportunities](#). Cancer Cell 27 (2015), pp. 382-396
  - **Cancer Driver Actionability Database (2014.12)**
    - Drug Interactions data in Cancer Drivers Database
    - Rubio-Perez, C., Tamborero, D., Schroeder, MP., Antolín, AA., Deu-Pons, J., Perez-Llamas, C., Mestres, J., Gonzalez-Perez, A., Lopez-Bigas, N. [In silico prescription of anticancer drugs to cohorts of 28 tumor types reveals novel targeting opportunities](#). Cancer Cell 27 (2015), pp. 382-396
  - **TCGA pan-cancer12 high confidence drivers (2013)**
    - doi:[10.1038/srep02650](https://doi.org/10.1038/srep02650)
  - **Cancer driver database (2013)**
    - doi:[10.1038/nmeth.2642](https://doi.org/10.1038/nmeth.2642)
  - **IntOGen Arrays (2010)**
    - doi:[10.1038/nmeth0210-92](https://doi.org/10.1038/nmeth0210-92).

# Analysis Page

- Log in to run your own mutations list through the IntOGen pipeline by uploading a mutations file.
  - Make sure file is formatted to specifications in link

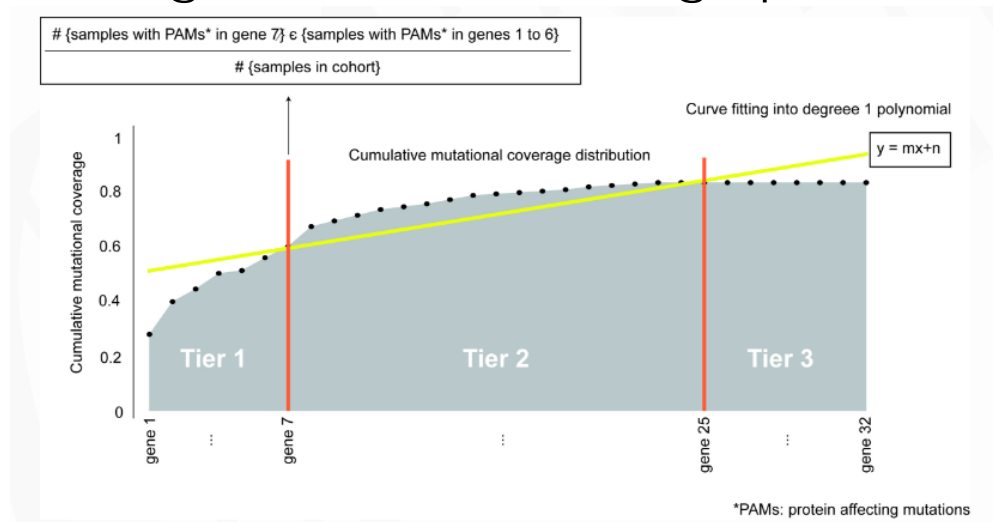


The screenshot shows the 'Cohort analysis' section of the IntOGen web interface. It features a blue header bar with a dropdown arrow. Below the header, there are three main input fields: 'Analysis name' with a text box and a description 'A name to identify easily your analysis.'; 'Mutations file' with a text box, a 'Browse ...' button, and a link 'this format' highlighted by a blue box; and 'Genome assembly' with a dropdown menu showing 'hg19 (GRCh37)'. At the bottom, there is a blue bar with 'Advanced options' and a right arrow, followed by a checkbox for 'Check if you understand and agree our Terms of Use.' and a 'Submit' button.



# OncoPaD

- <http://www.intogen.org/oncopad>
- Resource specifically designed to help design cost-effective NGS panels
  - Maximizing sample coverage
  - Minimizing the amount of DNA to sequence – focus on mutation hotspots, regulatory domains
- Need Google Account to design panel



# Design Page – Select Cancer type cohort

Panel name

Select cancer type cohort

☐ Select one or more cancer types to design a panel

☐ **H** Acute lymphocytic leukemia  
☐ **H** Acute myeloid leukemia  
☐ **GS** Bladder carcinoma  
☐ **S** Breast carcinoma  
☐ **H** Chronic lymphocytic leukemia  
☐ **S** Cutaneous melanoma  
☐ **S** **DS** Colorectal adenocarcinoma  
☐ **H** Diffuse B cell lymphoma  
☐ **S** **DS** Esophageal carcinoma  
☐ **S** **CNS** Glioblastoma multiforme  
☐ **S** **DS** Hepatocarcinoma  
☐ **S** Head and neck squamous cell carcinoma  
☐ **S** **CNS** Lower grade glioma  
☐ **S** **L** Lung adenocarcinoma

☐ **S** **L** Lung squamous cell carcinoma  
☐ **S** **CNS** Medulloblastoma  
☐ **H** Multiple myeloma  
☐ **S** **CNS** Neuroblastoma  
☐ **S** **L** Non small cell lung carcinoma  
☐ **S** **GS** Serous ovarian adenocarcinoma  
☐ **S** **CNS** Pilocytic astrocytoma  
☐ **S** **DS** Pancreas adenocarcinoma  
☐ **S** **GS** Prostate adenocarcinoma  
☐ **S** **GS** Renal clear cell carcinoma  
☐ **S** **L** Small cell lung carcinoma  
☐ **S** **DS** Stomach adenocarcinoma  
☐ **S** Thyroid carcinoma  
☐ **S** **GS** Uterine corpus endometrioid carcinoma  
☐ **S** Cervical

☒ Select one or more multi-cancer groups to design a panel

☒ **CNS** Central Nervous System malignancies  
☐ **DS** Digestive System carcinomas  
☐ **GS** Genitourinary System malignancies  
☐ **H** Haematological malignancies  
☐ **L** Lung carcinomas  
☐ **S** Solid tumors

☐ Consider your own cancer cohort ⓘ

Select cohort cancer type(s)

Mandatory to select one ⓘ

Format: VEP mutational output ⓘ

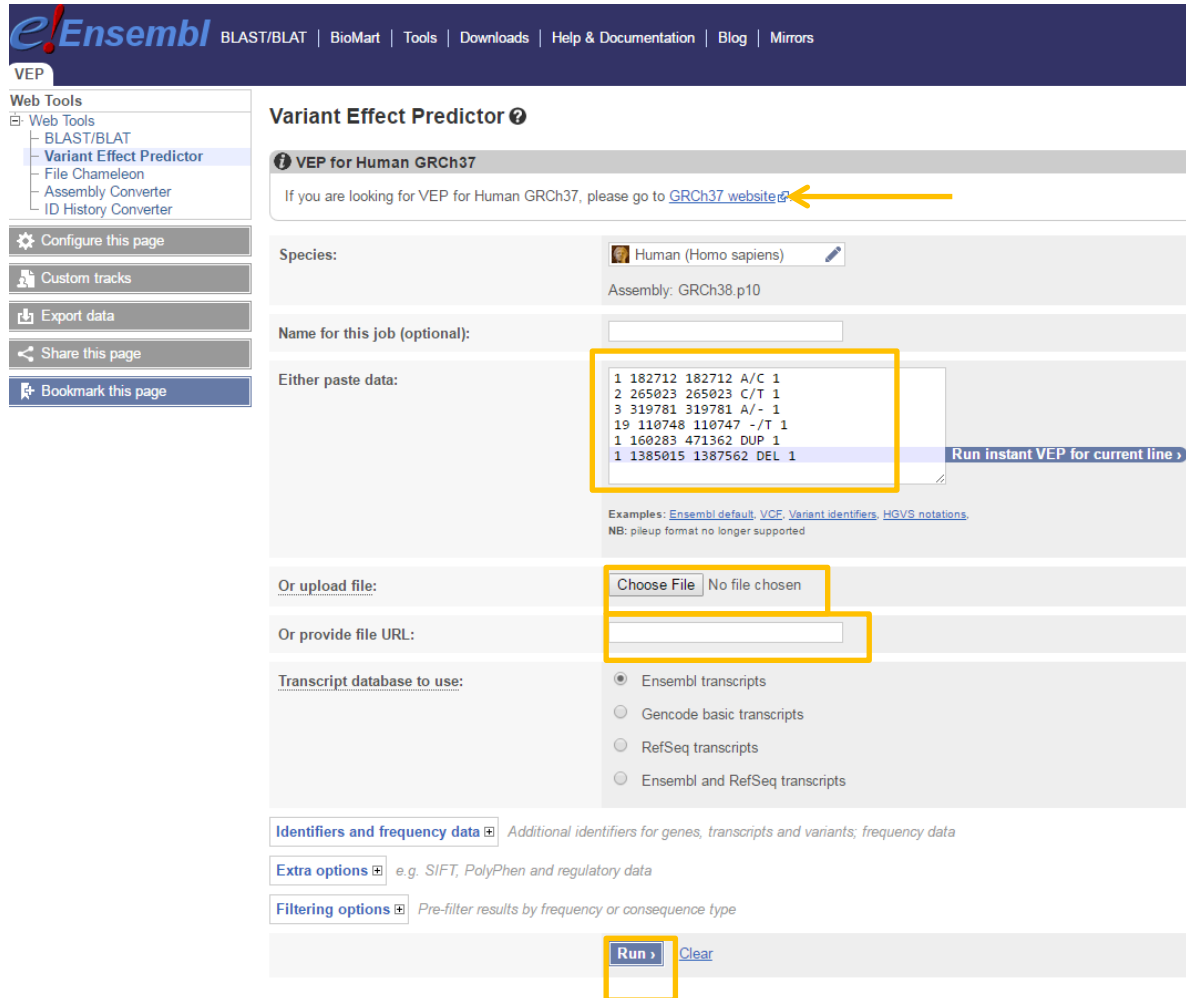
Example file

- 28 cancer types to pick from in top menu
  - Can select multiple options in this menu
- 6 multi-cancer group to pick from in bottom menu
  - Can select multiple options in this menu
- Or Consider your own cancer cohort
- You can only select one menu from which to select a cohort.

# Consider your own cancer cohort

- Select cohort cancer type from drop down list.
- Create/input VEP (Variant Effect Predictor) mutation file
  - TXT file generated from <http://www.ensembl.org/Tools/VEP>
    - To generate file on Ensembl site, you must use one of the following formats for your variants
      - Ensembl default: 1 1385015 1387562 DEL 1
      - VCF: 1 1385015 sv2 . <DEL> . . SVTYPE=DEL;END=1387562
      - Variant identifiers: RCV0000004642
      - HGVS notations: LRG\_101t1:c.1019T>C

# Creating VEP file



**Variant Effect Predictor**

VEP for Human GRCh37

If you are looking for VEP for Human GRCh37, please go to [GRCh37 website](#)

Species:  Assembly: GRCh38.p10

Name for this job (optional):

Either paste data:

1	182712	182712	A/C	1
2	265023	265023	C/T	1
3	319781	319781	A/-	1
19	110748	110747	-/T	1
1	160283	471362	DUP	1
1	1385015	1387562	DEL	1

[Run instant VEP for current line](#)

Examples: [Ensembl default](#), [VCF](#), [Variant identifiers](#), [HGVS notations](#).  
NB: pileup format no longer supported

Or upload file:  No file chosen

Or provide file URL:

Transcript database to use:

- ☒ Ensembl transcripts
- ☐ Gencode basic transcripts
- ☐ RefSeq transcripts
- ☐ Ensembl and RefSeq transcripts

Identifiers and frequency data Additional identifiers for genes, transcripts and variants; frequency data

Extra options e.g. SIFT, PolyPhen and regulatory data

Filtering options Pre-filter results by frequency or consequence type

- Can paste, upload mutations file or provide file URL to create VEP
- Select correct gene transcript set
- Make sure you are using the correct genome build (hg37 vs hg38)
- Select “Run”
- Save resulting file to input into design pad in OncoPaD.



# OncoPaD design page – Select candidate genes

Select candidate genes

☐ Include all driver genes (n=576) of the selected tumor type(s) and biomarkers of drug effect ⓘ

☐ Include only drug effect biomarker genes (n=297) ⓘ

☒ Consider your own list of genes (Max. 1000 genes)

Mandatory Validate to Submit

Format: Gene symbols comma separated

Fine tune the genes/regions to include in the panel

☐ Check if you understand and agree to our [Terms of Use](#).

Format: one gene symbol per line ⓘ

[Example file](#)

- Select candidate genes for panel.
  - All driver genes and biomarkers of drug effect for selected tumor type
  - Only drug effect biomarkers
  - Custom list of genes
    - Validate gene symbols from your list prior to uploading
  - Check box for understanding Terms of Use
  - Click 'Submit'

# OncoPaD design page – Advanced Settings

- Specify panel size
- Include/Exclude panel gene candidates
- Fine tune panel tier gene classification (See graph on slide 13)
  - Tier 1: Contribute most to cumulative mutational coverage
  - Tier 2: Contribute less to cumulative mutational coverage
  - Tier 3: Genes do not contribute to cumulative mutational coverage
- Fine tune gene regions parameters
- See screenshot on next page.

## ▼ Specify a panel size

Maximum number of genes to include in the panel ⓘ e.g. 20 (not limited by default)


Maximum Kpbs of DNA to sequence in the panel ⓘ e.g. 100 (not limited by default)

## ▼ Include/exclude panel gene candidates

\*Only if one of the pre-compiled gene lists was selected as input

Specify if you want to force **including one/several genes** into the panel candidates ⓘ

e.g. BRAF,TP53,KRAS,...


 Upload gene list

**File format:** one gene symbol per line

**Format:** gene symbols comma separated

Specify if you want to force **excluding one/several genes** from the panel candidates ⓘ

e.g. JAK1,JAK2,...

 Upload gene list

**File format:** one gene symbol per line

**Format:** gene symbols comma separated

## ▼ Fine tune panel tier gene classification

In this section you can modify the gene tier classification method. ⓘ

☐ Tick to consider **Tier 1 stringent classification**.


## ▼ Fine tune gene regions parameters

In this section you can fine-tune whether genes are included in full or by specific regions of high mutation density in the panel.

At least  of the mutations in a gene are demanded to be allocated in mutation high density regions, modify this parameter here. ⓘ

Specify if some genes should be included as whole genes and not regions in the panel (by default only TP53 is included whole gene): ⓘ

TP53

 Upload gene list

**File format:** one gene symbol per line

**Format:** gene symbols comma separated

☐ Tick to include all genes in the panel as **whole genes**, do not consider gene regions.

# Results page

Can download results in different formats

BED file

Print/PDF report

Excel file

My Panel was designed to interrogate Glioblastoma multiforme (GBM), Lower grade glioma (LGG), Medulloblastoma (MB), Neuroblastoma (NB) and Pilocytic astrocytoma (PA) tumors. 141 genes were in the input to generate the panel.

My Panel (only Tier 1 genes) would contain 45.88 DNA Kbps and include 19 genes/gene fragments; it would identify 45.0% of the 1069 tumors in the cohort.

## My Panel summary

Cancer Types

GBM, LGG, MB, NB, PA

Samples available for analysis

1069

### Tier 1 genes/regions 1

number of genes/regions

19

maximum mutational coverage

45.0%

### Tier 2 genes/regions 1

number of genes/regions

65

maximum mutational coverage

60.0%

Can view gene plots in graph format  
or all genes in table format

## My Panel mutational coverage

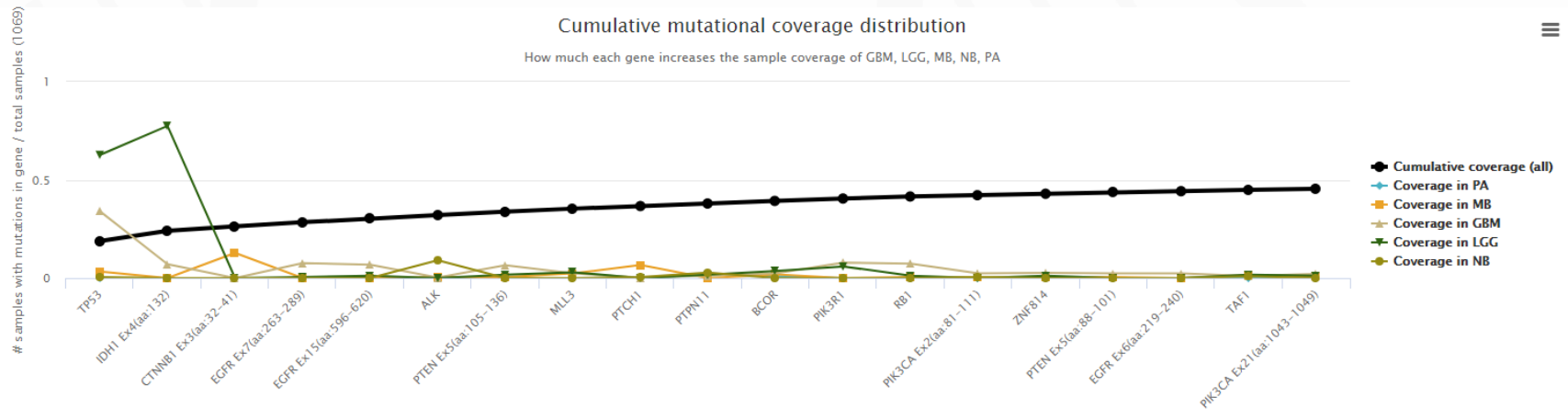
Tier 1 cumulative plot

Tier 2 cumulative plot

Table



### Cumulative mutational coverage distribution

How much each gene increases the sample coverage of GBM, LGG, MB, NB, PA



# Panel Mutational Coverage




**My Panel mutational coverage**

Tier 1 cumulative plot  Tier 2 cumulative plot  **Table**

Show **10** entries Search:

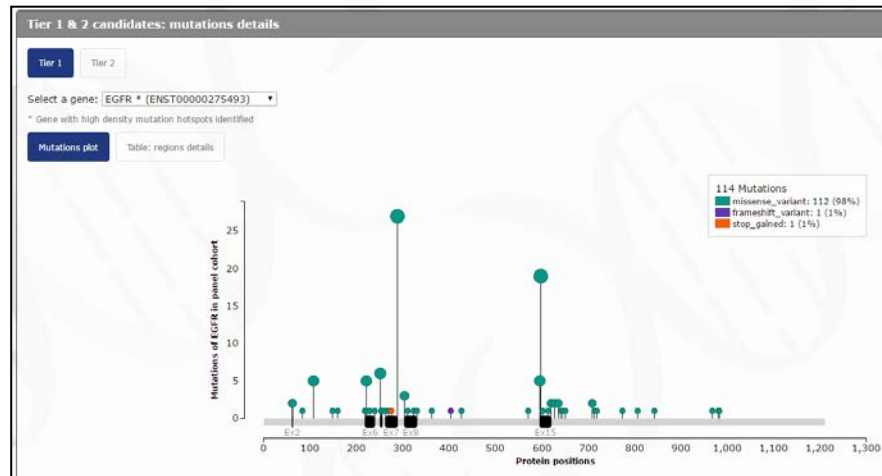
Gene/region	Panel tier	Bp length gene/region	Cumulative mutational coverage	Cumulative KBps in panel	Relevant mutations	GBM coverage	LGG coverage	MB coverage	NB coverage	PA coverage
TP53	Tier1	1182	0.188	1.182	Resp Resis	0.34	0.627	0.033	0.005	0.0
IDH1 Ex4(aa:132)	Tier1	2	0.24	1.184	Resp	0.071	0.775	0.0	0.0	0.0
CTNNB1 Ex3(aa:32-41)	Tier1	28	0.263	1.212	Onco Resis	0.0	0.0	0.129	0.0	0.0
EGFR Ex7(aa:263-289)	Tier1	80	0.284	1.292		0.077	0.006	0.0	0.0	0.0
EGFR Ex15(aa:596-620)	Tier1	75	0.302	1.367		0.069	0.012	0.0	0.0	0.0
ALK	Tier1	4863	0.321	6.23	Onco Resis	0.003	0.0	0.005	0.09	0.0
PTEN Ex5(aa:105-136)	Tier1	95	0.339	6.325	Resp Resis	0.066	0.018	0.005	0.0	0.0
MLL3	Tier1	14736	0.354	21.061		0.024	0.03	0.024	0.0	0.0
PTCH1	Tier1	4344	0.367	25.405	Resp	0.003	0.0	0.067	0.005	0.0
PTPN11	Tier1	1782	0.38	27.187		0.013	0.018	0.0	0.029	0.02

Showing 1 to 10 of 132 entries Previous **1** 2 3 4 5 ... 14 Next

 Onco Validated oncogenic  Resp Drug responsive  Resis Drug resistant

\* The next collapsed table below this one is the panel mutational coverage considering more than one gene per sample.

# Tier 1 & 2 candidates: mutations details



- Mutations observed per panel gene in visual gene transcript with mutation markers
- Table with mutations described in more detail including genomic coordinates

Tier 1 & 2 candidates: mutations details

Select a gene: **EGFR \* (ENST00000275493)**

\* Gene with high density mutation hotspots identified

Mutations plot

Table: regions details

Show **10** entries

Search: **EGFR**

Gene	Exon	Protein coords	Bp length	Mutations count	Mutations proportion	CDS proportion	Genomic coords
EGFR	Ex2	62-63	3	4	0.042	0.001	chr7:55210075-55210077
EGFR	Ex8	304-331	82	6	0.063	0.023	chr7:55223543-55223624
EGFR	Ex7	252-256	13	8	0.084	0.004	chr7:55221710-55221722
EGFR	Ex6	219-240	65	9	0.095	0.018	chr7:55220265-55220329
EGFR	Ex15	596-620	75	28	0.295	0.021	chr7:55233036-55233110
EGFR	Ex7	263-289	80	30	0.316	0.022	chr7:55221743-55221822

Showing 1 to 6 of 6 entries (filtered from 44 total entries)

Previous **1** Next

# Tier 1 & 2 candidates: relevant mutations details

Gene/region	Panel tier	Mutation	Mutation type	Drugs	Number of mutated tumors	Fraction of mutated tumors
ALK	Tier1	ALK R1275Q	Oncogenic		7	0.333
ALK	Tier1	ALK F1174L	Oncogenic		6	0.286
CTNNB1 Ex3(aa:32-41)	Tier1	CTNNB1 D32Y	Oncogenic		3	0.115
ALK	Tier1	ALK F1174C	Oncogenic		2	0.095
ALK	Tier1	ALK I1171N	Oncogenic		1	0.048
PTEN Ex8(aa:315-336)	Tier2	PTEN oncogenic mutation	Drug responsive	AKT inhibitors	18	0.947
PTEN Ex8(aa:315-336)	Tier2	PTEN oncogenic mutation	Drug responsive	Everolimus (MTOR inhibitor)	18	0.947
PTEN Ex8(aa:315-336)	Tier2	PTEN oncogenic mutation	Drug responsive	PARP inhibitors	18	0.947
PTEN Ex8(aa:315-336)	Tier2	PTEN oncogenic mutation	Drug responsive	PD1 Ab inhibitors	18	0.947
PTEN Ex8(aa:315-336)	Tier2	PTEN oncogenic mutation	Drug responsive	PI3K pathway inhibitor + AR antagonists	18	0.947

Showing 1 to 10 of 748 entries

Previous 1 2 3 4 5 ... 75 Next

- Click on magnifying glass or hover over mutation type to see details and/or references for the information in the table.
- Click over "i" in column headings to see description of information supplied in column

# Tier 1 & 2 candidates: general features

- Provides mode of action, supporting reference, and cancer (in your query) for which the gene has been identified

Gene	Panel tier	Driver mode of action	Driver source	Major driver
ALK	Tier1	Activating	Rubio-Perez&Tamborero(2015), CGC, Validated somatic mutation list	
BCOR	Tier1	Loss of function	Rubio-Perez&Tamborero(2015)	
MLL3	Tier1	Loss of function	Rubio-Perez&Tamborero(2015)	
PIK3R1	Tier1	Loss of function	Rubio-Perez&Tamborero(2015), CGC	
PTCH1	Tier1	No class	Rubio-Perez&Tamborero(2015), CGC	
PTPN11	Tier1	Activating	Rubio-Perez&Tamborero(2015)	
RB1	Tier1	Loss of function	Rubio-Perez&Tamborero(2015)	GBM
TAF1	Tier1	Activating	Rubio-Perez&Tamborero(2015)	
TP53	Tier1	Loss of function	Rubio-Perez&Tamborero(2015), CGC, Validated somatic mutation list	LGG,GBM
ZNF814	Tier1	Activating	Rubio-Perez&Tamborero(2015)	

Showing 1 to 10 of 54 entries

Previous 1 2 3 4 5 6 Next



# OncoPaD Literature

- <https://www.ncbi.nlm.nih.gov/pubmed/25759023>
- <https://genomemedicine.biomedcentral.com/articles/10.1186/s13073-016-0349-1>

# Scenario #1

- You are interpreting an NGS panel for bladder cancer and have encountered the missense mutations below. Which of these are driver mutations according to IntOGen?
  - FBXW7 T305K
  - TP53 R248K
  - ARID1A Q2188H
  - FGFR3 G237D

# Scenario #1

- Select the criteria "Bladder carcinoma" and "Gene of interest"
- Scroll down to "Mutations distribution along protein sequence" and toggle to Table view.
- Find variant of choice and click on "Driver classification" column
- FBXW7 T305K - Driver
- TP53 R248K - Driver
- ARID1A Q2188H - Passenger
- FGFR3 G237D – No data available

Mutations distribution along protein sequence

Choose a transcript: FGFR3-001 (807 aa) [Download] [Share]

Show 10 entries

Variant Locus	Samples	AA pos	AA change	Consequence	Driver classification
4:1803568:+:C/G	4	249	S/C	missense_variant	known in: BLCA;CER
4:1806119:+:G/A	2	380	G/R	missense_variant	predicted driver: tier 1
4:1808937:+:C/-	1	790	-	frameshift_variant	predicted passenger
4:1807559:+:C/T	1	576	-	synonymous_variant	not protein-affecting
4:1803564:+:C/T	1	248	R/C	missense_variant	known in: LUAD
4:1803435:+:G/A	1	235	G/D	missense_variant	predicted passenger
4:1803395:+:G/A	1	222	D/N	missense_variant	predicted driver: tier 1
4:1803377:+:G/A	1	216	E/K	missense_variant	predicted driver: tier 1

Showing 1 to 8 of 8 entries

Previous 1 Next

# Scenario #2

- You are creating an NGS panel for Bladder cancer.
  - Use OncoPaD tool to find candidate NGS targets.

# Scenario #2: Panel Design

- Select Bladder carcinoma as your target cancer type cohort.
- Select candidate gene pool
  - All driver genes of the selected tumor type and biomarkers of drug effect
- Under Advanced Settings – can specify a maximum number of genes/regions you want in your panel if applicable.

# Scenario #2: Results

- 16 regions were identified as either Tier 1 or Tier 2 mutation regions.

My Panel summary	
<b>Cancer Types</b>	BLCA
<b>Samples available for analysis</b>	98
<b>Tier 1 genes/regions</b> ⓘ	
number of genes/regions	5
maximum mutational coverage	85.0%
<b>Tier 2 genes/regions</b> ⓘ	
number of genes/regions	11
maximum mutational coverage	99.0%

My Panel was designed to interrogate Bladder carcinoma (BLCA) tumors. 159 genes were in the input to generate the panel.

The total number of genes included in the panel was set by user to 25.

My Panel (only Tier 1 genes) would contain 36.1 DNA Kbps and include 5 genes/gene fragments; it would identify 85.0% of the 98 tumors in the cohort.

# NGS Panel Creation Results

## Tier 1 Genes/Regions

- TP53
- MLL2
- ARID1A
- KDM6A
- EP300
- Green = information regarding gene mutations and response to therapy

## Tier 2 Genes/Regions

- FGFR3 – exon 7
- NUP107
- MLL3
- ZNF814 – exon 3
- CUL2
- BCLAF1
- PCDH18
- TSC1
- SMC1A
- CEP290
- GPS2

# Scenario #3

- Your institution is creating a list of genomic HotSpots for mutations (SNVs)
  - All oncology samples
  - Specific diseases (i.e. AML)
- Use “Catalog of driver mutations” to navigate driver mutations seen frequently in cancer.



# Scenario 3A

- Pan Cancer Mutation HotSpots
- From homepage scroll down to “Catalog of driver mutations” section and toggle to table view.
- Determine appropriate percentage of samples with mutations to add to list.
- Log in to download list as .csv

# Scenario 3B

- AML Mutation HotSpots
- Select 'AML – Acute Myeloid Leukemia' from the search bar
- Scroll down to "Catalog of driver mutations" and toggle to table view.
  - Visible list will include all mutations seen in at least 4 samples, but downloaded list will include more.

# Contact Information

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