Intro to cBioPortal

1. Recent Progress

2. ITCR Collaborations

3. Plans
Intro to cBioPortal
Web platform that enables researchers to **visualize** and **analyze** large-scale cancer genomic data sets.

**Focussed on ease of use;** engaging biologists and translational researchers.
Fully open source and developed across five groups.

MSKCC, DFCI, Children’s Hospital of Philadelphia, Princess Margaret Cancer Centre, and The Hyve.
Available as a public site: cbioportal.org

Increasingly being used within cancer centers via local installations.
2018: 210K users
2019: 118K users so far (~235K projected)

>4,000 citations
The cBioPortal for Cancer Genomics provides visualization, analysis and download of large-scale cancer genomics data sets. Please adhere to the TCGA publication guidelines when using TCGA data in your publications.

### Uterine Corpus Endometrial Carcinoma (TCGA, Provisional)

Samples with mutation and CNA data (242 patients/samples) - ERBB2, KRAS & 4 other genes

Queried genes are altered in 221 (91%) of queried patients/samples

<table>
<thead>
<tr>
<th>Gene</th>
<th>Alteration</th>
</tr>
</thead>
<tbody>
<tr>
<td>ERBB2</td>
<td>12%</td>
</tr>
<tr>
<td>KRAS</td>
<td>22%</td>
</tr>
<tr>
<td>NRAS</td>
<td>5%</td>
</tr>
<tr>
<td>PIK3CA</td>
<td>57%</td>
</tr>
<tr>
<td>PIK3R1</td>
<td>34%</td>
</tr>
<tr>
<td>PTEN</td>
<td>67%</td>
</tr>
</tbody>
</table>

**Genetic Alteration**
- Inframe Mutation (putative driver)
- Inframe Mutation (unknown significance)
- Missense Mutation (putative driver)
- Missense Mutation (unknown significance)
- Truncating Mutation (putative driver)
- Truncating Mutation (unknown significance)
- Amplification
- Deep Deletion
- No alterations

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**Main Results Page**
### 60 Mutations (page 1 of 6)

<table>
<thead>
<tr>
<th>Gene</th>
<th>Protein Change</th>
<th>Annotation</th>
<th>Mutation Type</th>
<th>Allele Freq</th>
<th>Copy #</th>
<th>Cohort</th>
<th>COSMIC</th>
</tr>
</thead>
<tbody>
<tr>
<td>AKT1</td>
<td>E17K</td>
<td></td>
<td>Missense</td>
<td>0.42</td>
<td>Gain</td>
<td>4.2%</td>
<td>283</td>
</tr>
<tr>
<td>TP53</td>
<td>V274F</td>
<td></td>
<td>Missense</td>
<td>0.36</td>
<td>ShallowDel</td>
<td>34.3%</td>
<td>78</td>
</tr>
<tr>
<td>GATA3</td>
<td>X350_splice</td>
<td></td>
<td></td>
<td>0.24</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ETV4</td>
<td>R134Gfs*49</td>
<td></td>
<td>Splice</td>
<td>0.16</td>
<td>Diode</td>
<td>11.8%</td>
<td></td>
</tr>
<tr>
<td>PTPRD</td>
<td>T342M</td>
<td></td>
<td>Missense</td>
<td>0.38</td>
<td>ShallowDel</td>
<td>0.2%</td>
<td></td>
</tr>
<tr>
<td>ACTL6B</td>
<td>N233Kfs*17</td>
<td></td>
<td>Missense</td>
<td>0.13</td>
<td></td>
<td>1.2%</td>
<td>1</td>
</tr>
<tr>
<td>JPH4</td>
<td>A502Gfs*12</td>
<td></td>
<td></td>
<td>0.11</td>
<td></td>
<td>0.7%</td>
<td>27</td>
</tr>
<tr>
<td>GLYR1</td>
<td>R381Afs*15</td>
<td></td>
<td></td>
<td>0.10</td>
<td></td>
<td>1.2%</td>
<td>3</td>
</tr>
<tr>
<td>PAX2</td>
<td>V266Gfs*28</td>
<td></td>
<td></td>
<td>0.13</td>
<td></td>
<td>0.5%</td>
<td></td>
</tr>
<tr>
<td>LIN28A</td>
<td>A17E</td>
<td></td>
<td>Missense</td>
<td>0.29</td>
<td></td>
<td>0.1%</td>
<td></td>
</tr>
</tbody>
</table>

### 690 Copy Number Alterations (page 1 of 69)

<table>
<thead>
<tr>
<th>Gene</th>
<th>CNA</th>
<th>Annotation</th>
<th>Cytoband</th>
<th>Cohort</th>
</tr>
</thead>
<tbody>
<tr>
<td>PIK3CA</td>
<td>AMP</td>
<td></td>
<td>3q28.32</td>
<td>0.4%</td>
</tr>
<tr>
<td>TERT</td>
<td>AMP</td>
<td></td>
<td>5p15.33</td>
<td>0.1%</td>
</tr>
<tr>
<td>CCND1</td>
<td>AMP</td>
<td></td>
<td>11q13.3</td>
<td>16.2%</td>
</tr>
<tr>
<td>CCNE1</td>
<td>AMP</td>
<td></td>
<td>19q12</td>
<td>4.2%</td>
</tr>
</tbody>
</table>

Patient View Page
cBioPortal is now installed locally at ~40 sites.
Recent Progress
Architecture Upgrade

- JSON-based REST API
- Testable code (less buggy)
- Continuous integration
- Better performance and scalability
- Easier to maintain and add new features
Revised Web API

- JSON-based REST web API, redesigned based on Open API specification
- Complete access to all data in cBioPortal
- Greater interoperability with other tools
- API clients for Java, Python, Javascript, R, and other languages (*work in progress*)
- Token-based API access for private instances of cBioPortal (*work in progress*)
Education and Outreach

- **Resources**
  - Tutorial slides
  - Updated FAQ
  - Email support via google groups

- **User Trainings**
  - Offered regularly in Boston & NYC
  - Online training coming soon

- **Google Summer of Code**
  - 6 students participating this year

- **Conference Presentations**
  - Talk at AACR Annual Meeting
Quick Search

<table>
<thead>
<tr>
<th>Study Title</th>
<th>Samples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Brain Lower Grade Glioma (TCGA, Provisional)</td>
<td>530</td>
</tr>
<tr>
<td>Brain Lower Grade Glioma (TCGA, PanCancer Atlas)</td>
<td>514</td>
</tr>
<tr>
<td>Breast Invasive Carcinoma (TCGA, Cell 2015)</td>
<td>817</td>
</tr>
</tbody>
</table>

47 more studies (click to load 20 more)

<table>
<thead>
<tr>
<th>Gene</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>BRAF</td>
<td>7q34</td>
</tr>
<tr>
<td>BRAFP1</td>
<td>Xq13.3</td>
</tr>
<tr>
<td>BRAF_PS147</td>
<td>7q34</td>
</tr>
</tbody>
</table>

748 more genes (click to load 20 more)

<table>
<thead>
<tr>
<th>Patient</th>
<th>Study Title</th>
</tr>
</thead>
<tbody>
<tr>
<td>BR-M-005</td>
<td>Breast Invasive Carcinoma (Broad, Nature 2012)</td>
</tr>
<tr>
<td>BR-M-026</td>
<td>Breast Invasive Carcinoma (Broad, Nature 2012)</td>
</tr>
<tr>
<td>BR-M-027</td>
<td>Breast Invasive Carcinoma (Broad, Nature 2012)</td>
</tr>
</tbody>
</table>

793 more patients (click to load 20 more)
Virtual Studies

- Breast Cancer (MSK, Cancer Cell 2018)
- Breast Fibroepithelial Neoplasms (Duke-NUS, Nat Genet 2015)
- Breast Cancer Xenografts (British Columbia, Nature 2015)
- Breast Invasive Carcinoma (British Columbia, Nature 2012)
- Breast Invasive Carcinoma (Broad, Nature 2012)
- Breast Invasive Carcinoma (Sanger, Nature 2012)
- Breast Invasive Carcinoma (TCGA, Cell 2015)
Annotation tracks in Lollipop plots
ITCR Collaborations
Integrative Genomics Viewer (IGV)
Collaboration with Jim Robinson, Helga Thorvaldsdottir, and Jill Mesirov
Clinical Interpretation of Variants in Cancer (CIVIC)
Collaboration with Obi Griffith et al.
Next-Generation Clustered Heat Map (NG-CHM)
Collaboration with John Weinstein and Bradley Broom
Cancer Digital Slide Archive (CDSA)
Collaboration with David Gutman et al.
BioConductor Package (work in progress)
Collaboration with Levi Waldron and Marcel Ramos
Plans 04
Plans for Upcoming Year

- Cohort comparison
- Authentication support for API
- IGV.js in the patient view
- Waterfall and swimmers plots
- Single-cell support (via HTAN)
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Patient: TCGA-02-0006, Female, Glioblastoma Multiforme (Glioblastoma Multiforme), DECEASED (18 months), Recurred (9 months)
Samples: TCGA-02-0006-01, Primary

Summary | Clinical Data | Pathology Report | Tissue Image
---|---|---|---
| | | | 23.9%

CNA MUT
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