Introduction to cBioPortal

Welcome!

Course material

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Andrew Mason BSc (Hons) PhD AFHEA

York Against Cancer

Lecturer in Cancer Informatics at The University of York

- Run a small bioinformatics-focused research group within the Jack Birch Unit
- Work on human urothelial cancers and retroviral cancers in birds
- Bioinformatic lead for the bladder cancer group of the 100,000 genomes project
- Just started my undergraduate teaching role

Elixir-UK Data Stewardship Training Fellow

- Improve data management, not just analysis, in life sciences
- Development of data management training resources

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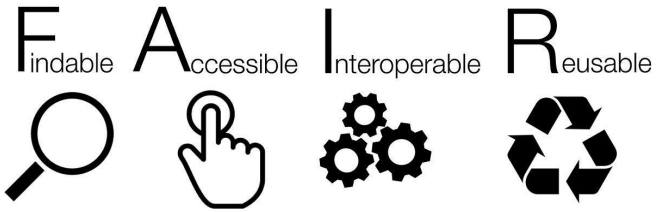
What is Elixir?

"ELIXIR coordinates and develops life science resources across Europe so that researchers can more easily find, analyse and share data, exchange expertise, and implement best practices."

Improve skills in data management

Improve quality of, and access to, informatics training

Develop and disseminate FAIR data principles in life sciences



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Elixir-UK Data Stewardship Training Fellows

Research Data Management bites → My videos introducing sequencing data

Online training courses, cookbooks and carpentries

Local, in-person training

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UK Research and Innovation

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Introduction and Learning Objectives

Introduction to cBioPortal

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Session structure

12.00	Introduction and Learning Objectives
12.10	cBioPortal website demonstration
12.20	Problem-solving tasks
12.45	Recap and Further Resources
12.50	Accessing and using underlying cBioPortal data
13.00	Close

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Learning objectives

1. Recognise the applications and utility of cBioPortal for cancer research
2. Operate and explore the cBioPortal website to identify cancer data of interest
3. Complete two cancer biology problem-solving tasks using cBioPortal
4. Recognise the process for accessing and analysing cBioPortal data

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What is cBioPortal?

cbioportal.org

Public website for exploratory analysis, visualisation and download of large cancer omics datasets, with clinical metadata

Data derived from large consortia, as well as highlighted studies

Data annotated by external reference databases

Depending on the dataset, includes mutations, CNA, gene expression, methylation data *etc.*

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When using cBioPortal, cite the following papers, plus the reference papers for datasets used: Cerami *et al.* 2012, Gao *et al.* 2013

Why use cBioPortal?

cBioPortal FOR CANCER GENOMICS

cbioportal.org

Access to the largest, publicly available cancer sequencing studies, all in one place

Explore broader relevance of laboratory/animal studies across cancer types

Hypothesis generation, including student projects

Data visualisation

Exploration of clinical data

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cBioPortal website demo

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Problem-solving tasks

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Task 1 – Exploration of the METABRIC breast cancer dataset

Task 2 – Exploration of two AML datasets

Task 3 – Exploration and comparison of two kidney cancers

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Recap and Further Resources

Introduction to cBioPortal

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Further training resources

cBioPortal Docs

cBioPortal | Github | Issues | Google Group

Filter

- Home
- About Us
- List of RFCs
- User Guide
  - New Users
  - Frequently Asked Questions
  - Overview of Resources
  - Page Specific Resources
  - OQL
  - News
  - Genie News
  - Web API and API Clients
- Deployment
- Development

Overview

Overview of Resources

Tutorial Slides

These tutorial slides contain annoted screenshots to walk you through using the cBioPortal site.

1. Single Study Exploration Google slides | PDF
2. Single Study Query Google slides | PDF
3. Patient View Google slides | PDF
4. Virtual Studies Google slides | PDF
5. Onco Query Language (OQL) Google slides | PDF
6. Group Comparison Google slides | PDF
7. Pathways Google slides | PDF

Webinar Recordings

Recordings of live webinars from April & May 2020

1. Introduction to cBioPortal youtube.com | bilibili.com | Download PDF | View slides
2. Mutation Details & Patient View youtube.com | bilibili.com | Download PDF | View slides
3. Expression Data Analysis youtube.com | bilibili.com | Download PDF | View slides
4. Group Comparison youtube.com | bilibili.com | Download PDF | View slides
5. API & R Client youtube.com | bilibili.com | Download PDF | View slides | Workshop code

How-To Videos

Short videos that show how to perform specific analyses or how to use specific pages.

Direct link

cBioPortal FAQs

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Accessing and using underlying cBioPortal data

Introduction to cBioPortal

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4 Recognise the process for accessing and analysing cBioPortal data

Why bother?

"under-the-hood" dataset has more information than displayed publicly

analyse lists of genes quickly

"improve" the plot quality

perform more advanced statistical testing (e.g. DEA, GSEA)

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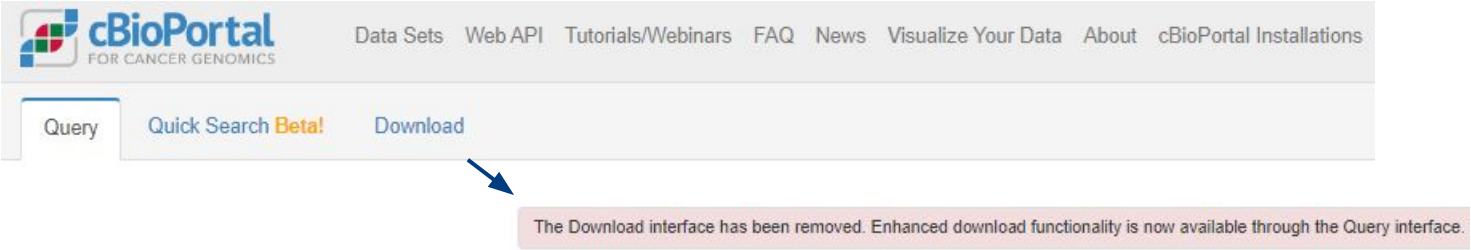
Downloading data



Unhelpful starting point.

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Downloading data

Explore your dataset first, and then download.

Download will start and give a `.tar.gz` file

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Downloading data

blca_tcga_pub_2017 | Search blca_tcga_pub_2017

Name	Date modified	Type	Size
case_lists	25/03/2022 19:07	File folder	
data_clinical_patient.txt	25/03/2022 19:15	TXT File	355 KB
data_clinical_sample.txt	25/03/2022 19:15	TXT File	103 KB
data_cna.txt	25/03/2022 19:15	TXT File	22,499 KB
data_linear_cna.txt	25/03/2022 19:15	TXT File	64,382 KB
data_methylation_hm450.txt	25/03/2022 19:15	TXT File	119,597 KB
data_mrna_seq_v2_rsem.txt	25/03/2022 19:15	TXT File	69,336 KB
data_mrna_seq_v2_rsem_zscores_ref_all_samples.txt	25/03/2022 19:15	TXT File	60,450 KB
data_mrna_seq_v2_rsem_zscores_ref_diploid_samples.txt	25/03/2022 19:15	TXT File	59,941 KB
data_mutations.txt	25/03/2022 19:15	TXT File	264,752 KB
data_mutsig.txt	25/03/2022 19:15	TXT File	2,083 KB
data_rppa.txt	25/03/2022 19:15	TXT File	643 KB
data_rppa_zscores.txt	25/03/2022 19:15	TXT File	569 KB
LICENSE	25/03/2022 19:07	File	1 KB
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meta_study.txt	25/03/2022 19:07	TXT File	1 KB

For each assay, 1 data file and 1 metadata/information file

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Downloading data

blca_tcga_pub_2017 | Search blca_tcga_pub_2017

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Understanding the data

data_clinical_patient.txt	25/03/2022 19:15	TXT File	355 KB
data_clinical_sample.txt	25/03/2022 19:15	TXT File	103 KB

TSV – feature x patient ID (many missing values, cancer-specific features)

Patient information

Sex, height, weight, race, ethnicity, diagnosis age, survival status

Occupation history, smoking status, family history

Tumour information

Stage, grade, disease codes, metastasis status

Tumour-specific categories (e.g. for bladder, rate of prostate cancer)

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Understanding the data

data_cna.txt	25/03/2022 19:15	TXT File	22,499 KB

tumour x gene using GISTIC scale (TSV)

- -2 homozygous "deep" deletion
- -1 shallow deletion (anything that isn't total loss)
- 0 diploid
- 1 gain ("a few" extra copies)
- 2 amplification (often in focal sets)

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Understanding the data

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tumour x gene, normalised gene expression data (TSV)

- Normalised counts
- Can feed into differential expression pipelines (DESeq2 *etc*), if careful!

- Good for comparisons of <u>one</u> gene across samples
- Harder to compare expression between genes of same sample

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Understanding the data

data_mutations.txt	25/03/2022 19:15	TXT File	264,752 KB

TSV – list of all mutations, sorted by tumour ID

- Includes synonymous mutations as well as non-synonymous
- Data structure is rubbish, requires lots of parsing to find hotspots *etc.*

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Working with the data

Existing UG training and extensive core bioinformatic support

Specific cBioPortal REST API for programmatic access

Python support available too – pandas package is versatile

Doable…! But. Memory intensive, and watch delimiters when importing.

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Working with the data… final thoughts

The data is not always complete

- Inconsistent column usage between datasets
- Watch 'whitespace' vs 'tab space' vs comma delimiters

Biological vs Statistical significance

Limited by previous bioinformatic analysis pipelines, genome version *etc.*

- More advanced questions can go back to the raw data

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Introduction to cBioPortal

Course complete!

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