CBIIT GigaGalaxy – A Galaxy-based Platform for Large-scale Genomics Analysis

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CBIIT



香港中文大學 - 華大基因研究所 跨組學創新研究院 CUHK-BGI Innovation Institute of Trans-Omics





- Jointly established between The Chinese University of Hong Kong (CUHK) and BGI.
- "We aim to provide a platform conducive to training of multi-disciplinary talents conversant with the knowledge and application of genomics, proteomics, genetics, computation biology and bioinformatics, by capitalizing on both institutions' expertise and strengths in genomic science."





Big Data Translates into Big Opportunities... and Big Responsibilities



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Nature 455, 461 (25 September 2008) | doi:10.1038/455461c; Published online 24 September 2008

Big data: open-source format needed to aid wiki collaboration

Tin-Lap Lee¹

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iklomics' (Nature 455, 22–25; 2008) points out that the yle community collaboration offers a smart way to respond a. Although lack of recognition and credit may prevent y participating, this may be only part of the story. Reasons the community can be more complicated.

g big data sets is becoming more and more challenging. is convenient, and the GenMAPP pathway database yumple. But the lack of a unified data format for facilitating e database to another can be a killer for someone trying to currently no de facto standard on pathway-data format, shes data portability.





The challenges for biomedical scientists







The challenges for biomedical scientists









Data intensive biology for everyone.

<u>Galaxy</u> is an open, web-based platform for data intensive biomedical research. Whether on the <u>free public server</u> or <u>your own instance</u>, you can perform, reproduce, and share complete analyses.

Use Galaxy



Use the free public server

Get Galaxy



Install locally or in the cloud

Learn Galaxy



Screencasts, Galaxy 101, ...

Get Involved



Mailing lists, Tool Shed, wiki

Search all resources

The <u>Galaxy Team</u> is a part of <u>BX</u> at <u>Penn State</u>, and the <u>Biology</u> and <u>Mathematics and Computer Science</u> departments at <u>Emory University</u>. The Galaxy Project is supported in part by <u>NHGRI</u>, <u>NSF</u>, <u>The Huck Institutes of the Life Sciences</u>, <u>The Institute for CyberScience at Penn State</u>, and <u>Emory University</u>.





CBIIT GigaGalaxy

Highlights:

- Provides enhanced functionality in additional to the original Galaxy functions
 - Specialized instances
 - Speed: local servers with SBS-UCSC genome database mirror in Hong Kong
 - Reproducibility: Seamless integration with Taverna/myExperiment workflows
 - > Data exchange and publishing: GigaScience journal portal/GigaDB
 - Customized functions and more.....





CBIIT GigaGalaxy

Benefits:

- ➤ Simplifies complicated bioinformatics tasks, accelerate data processing and allow flexible analysis.
- ➤ Significantly reduce software and hardware costs, encourage research collaboration.



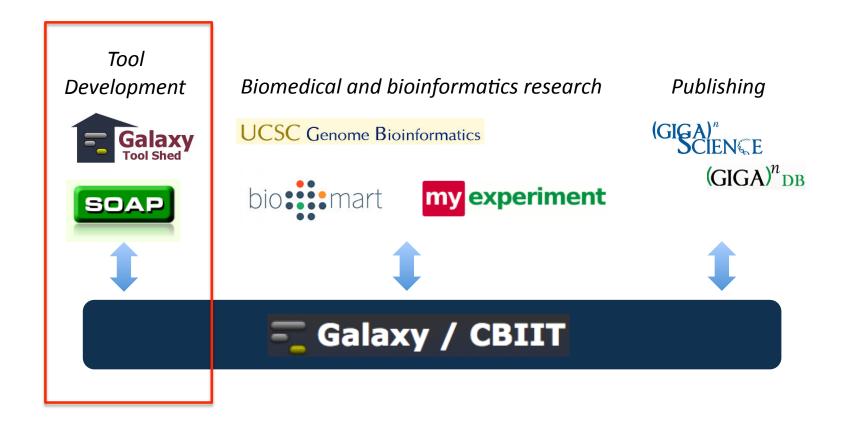








CBIIT GigaGalaxy Structure









What is SOAP?

• **SOAP** - a tool package that provides full solution to NGS data analysis by BGI.

| Soft | ware | | | |
|----------|---|--|--|--|
| • | SOAP3 is a GPU-based software for aligning short reads with a reference sequence. It can find all alignments with k mismatches where k is chosen from 0 to 3. When compared with its previous version SOAP2, SOAP3 can be up to tens of times faster. | | | |
| ✓ | SOAPaligner/soap2 | SOAPaligner/soap2 is a program for faster and efficient alignment for short oligonucleotide onto reference sequences. SOAPaligner/soap2 is compatible with numerous applications, including single-read or pairend resequencing. | | |
| • | SOAPsplice NEW | SOAPsplice is designed to use RNA-Seq reads for genome-wide ab initio detection of splice junction sites and identification of alternative splicing (AS) events. | | |
| V | SOAPsnp | SOAPsnp is an accurate consensus sequence builder based on soap1 and SOAPaligner/soap2's alignment output. It calculates a quality score for each consensus base, which can be used for any latter process to call SNPs. | | |
| V | SOAPdenovo | SOAPdenovo, a short read de novo assembly tool, is a package for assembling short oligonucleotide into contigs and scaffolds. | | |
| 4 | SOAPindel | SOAPindel is developed to find the insertion and deletion specially for re-sequence technology. | | |
| ✓ | SOAPsv | SOAPsv is a program for detecting the structural variation . | | |
| 1 | SOAP v1 | SOAP v1 is available all the same. | | |





Why SOAP?

- Galaxy has been using SAMtools for consensus sequence calling, but the recent upgrade has left this part out, which is very limited to some biologists.
- SOAPsnp is the only other method that can call full consensus sequences besides SAMtools.
- The main galaxy site supports none of the SOAP tools, including SOAPsnp.





Galaxy Tool Shed

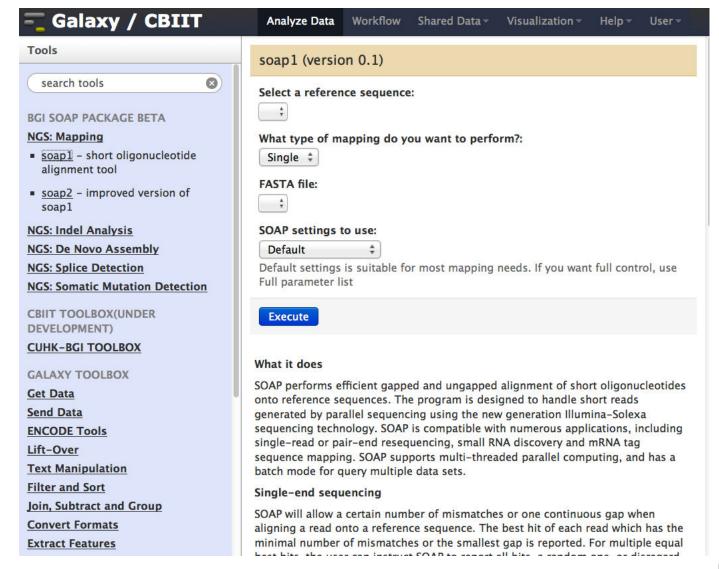
- Enables sharing of Galaxy tools across Galaxy servers around the world.
- SOAP package tools configured for use in Galaxy.
 - SOAPsnp/SOAPdenovo







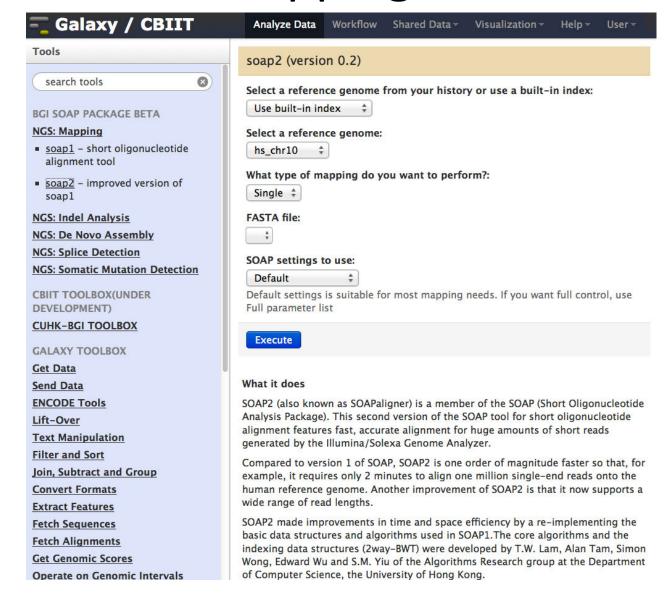
NGS mapping: SOAP1







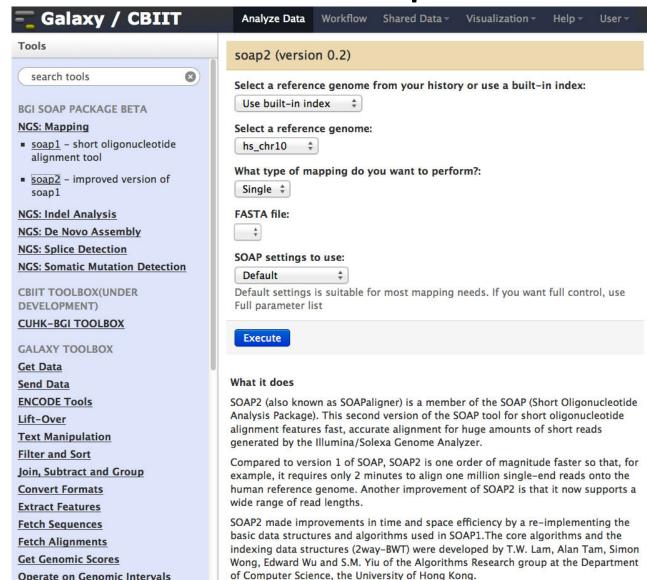
NGS mapping: SOAP2







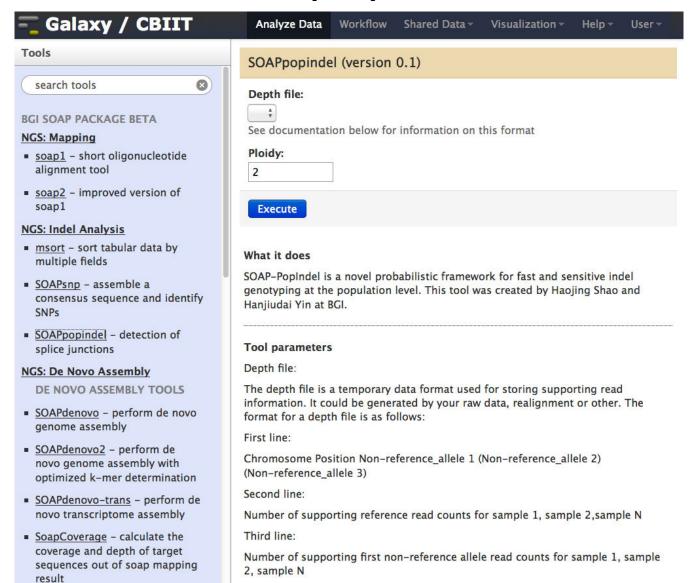
SOAPsnp







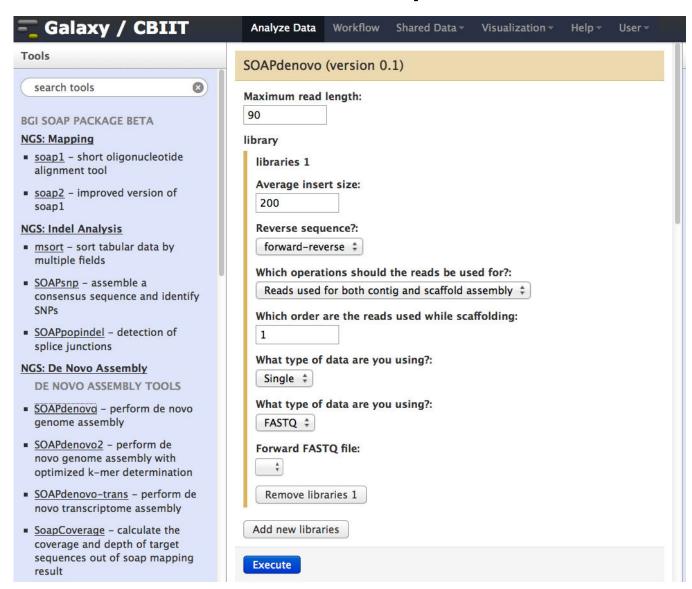
SOAPpopindel







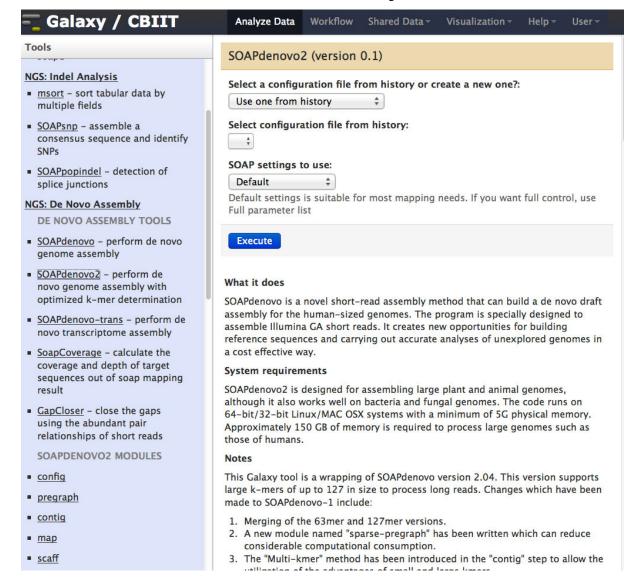
NGS De Novo Assembly: SOAPdenovo







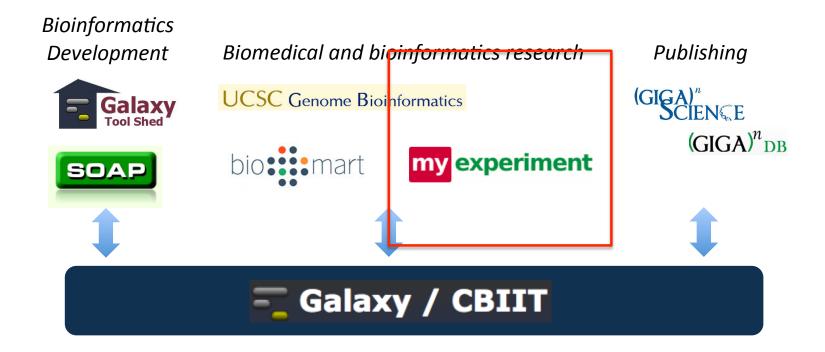
NGS De Novo Assembly: SOAPdenovo2







CBIIT GigaGalaxy structure







my experiment How does it work?



http://www.myexperiment.org

myExperiment -a repository for workflows.

> Taverna workflows.



➤ New: Galaxy workflows.



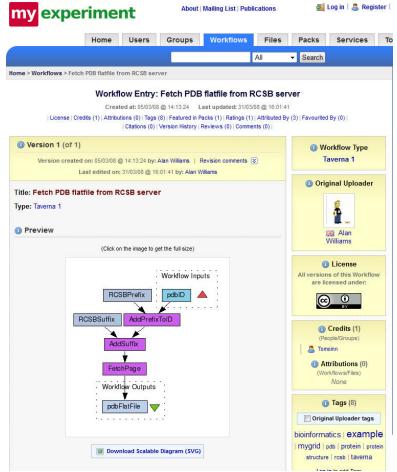
• **CBIIT GigaGalaxy** integration

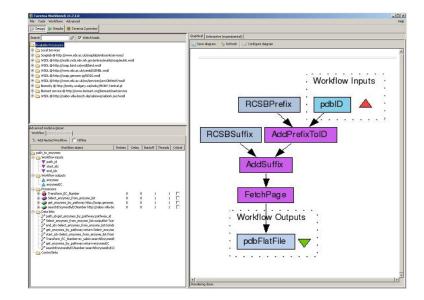






Taverna workflow

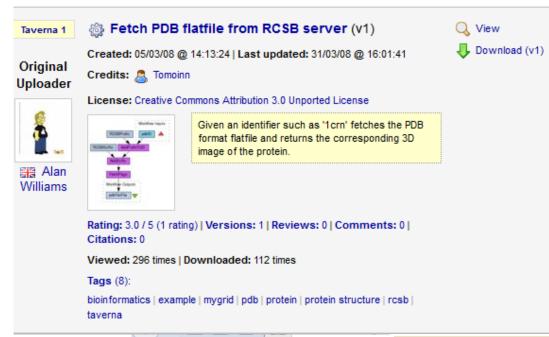














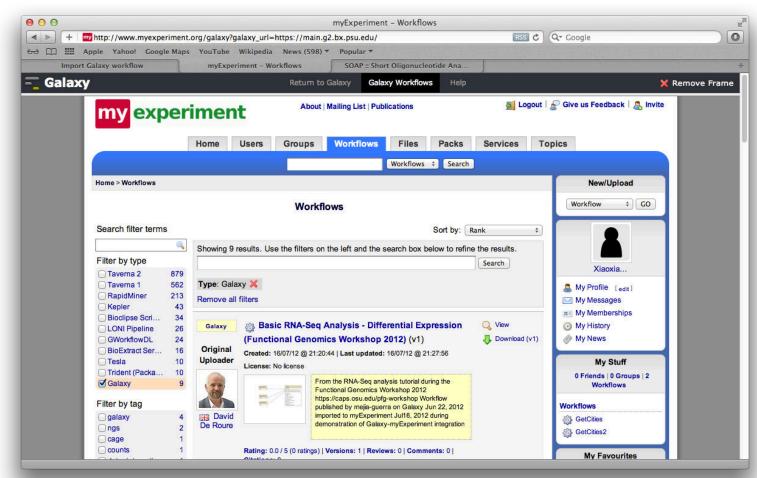


Inputs

pdbID PDB identifier such as '1crn' Examples include: 1crn

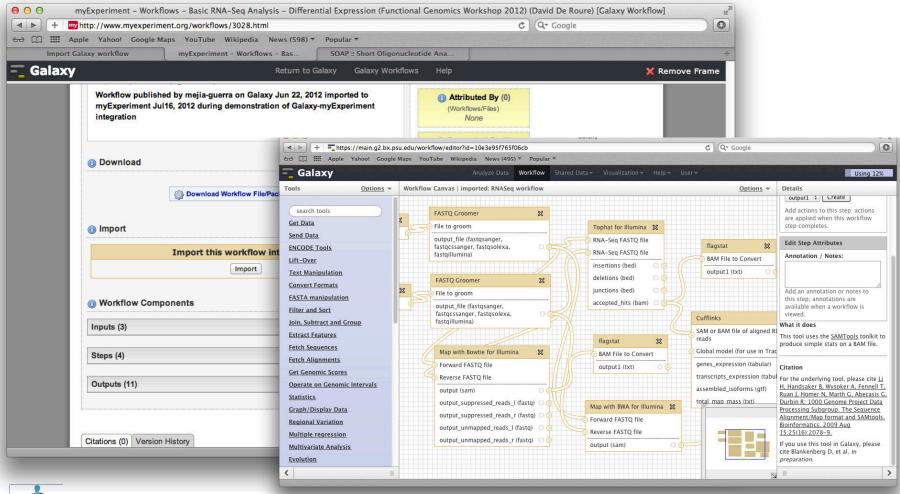


Galaxy workflow





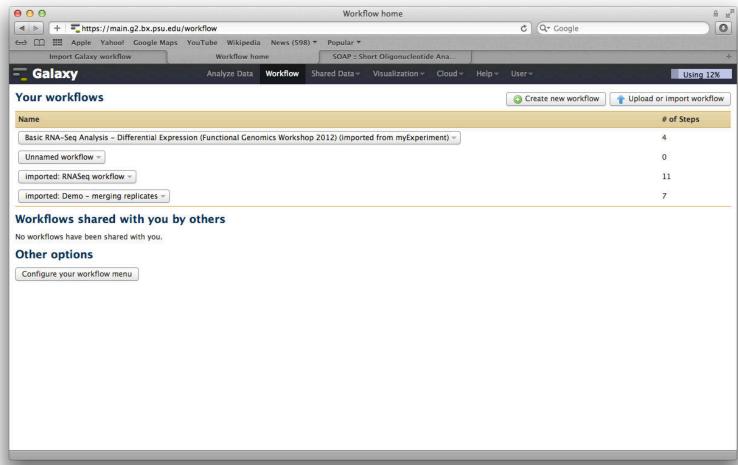
Import (1)







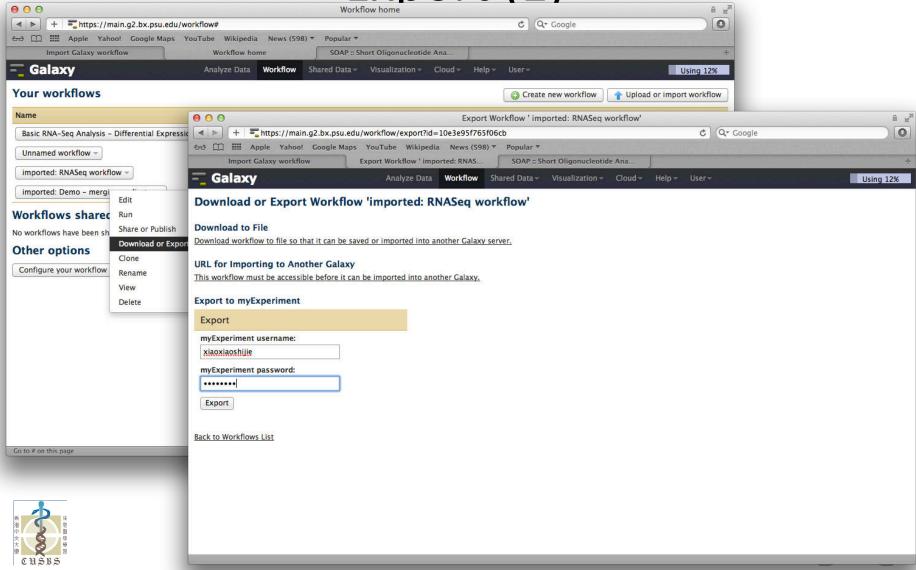
Import (2)



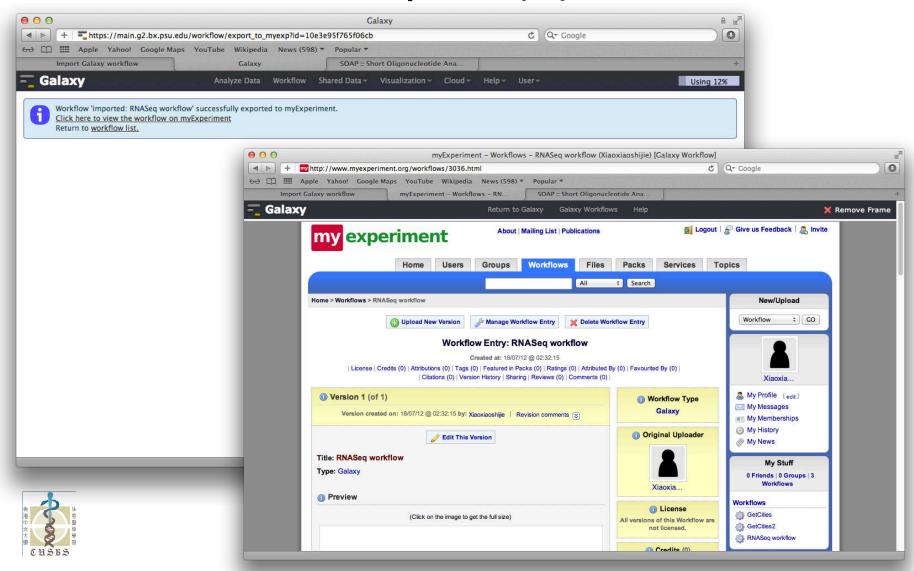




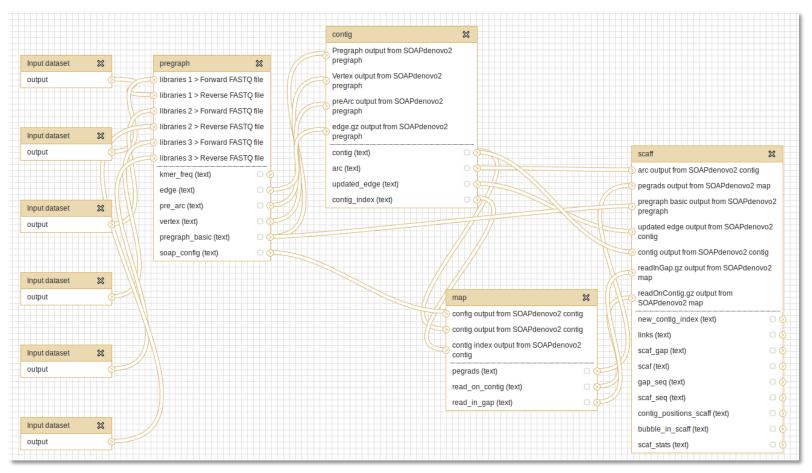
Export (1)



Export (2)



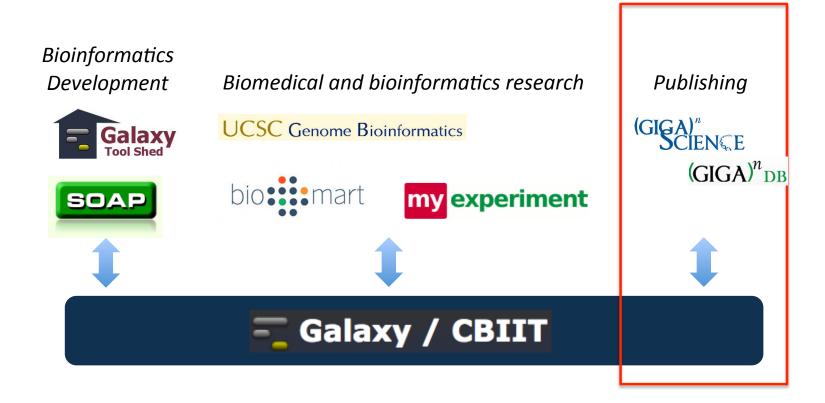
SOAPdenovo2 Galaxy workflow







CBIIT GigaGalaxy structure







IGGAN Now launched...
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LARGE-SCALE DATA JOURNAL/DATABASE



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Large and linked in scientific publishing

Laurie Goodman, Scott C Edmunds, Alexandra T Basford

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Abstract | Full text | PDF |

Editor's summary

Cochrane et al. GigaScience Preview http://www.gigasciencejournal.com



REVIEW

Onen Access

The future of DNA sequence archiving

Guy Cochrane*, Charles E Cook and Ewan Birney

Abstract

Archives operating under the International Nucleotide Sequence Database Collaboration currently preserve all submitted sequences equally, but rapid increases in the rate of global sequence sequally, but rapid increases in the rate of global sequence production will soon require differentiated treatment of DNA sequences submitted for archiving. Here, we provide a background on the establishment and operation of public data repositories and present the issues the community faces given the current overwhelming increase in data output. We also propose a way forward through the use a graded system in which the ease of reproduction of a sequencing-based experiment and the relative availability of a sample for resequencing be used as a means to define the level of lossy compression to the stored data.

Keywords DNA, sequence, archive, compression, storage, image

The vast majority of living organisms utilise nucleic acid as their primary store of genetic information. The technology to sequence DNA routinely was developed in the 1970s, but advances over time have since reduced cost and increased output. As the cost of sequencing has laboratory techniques in which DNA and RNA can be cut, ligated, interconverted and replicated in vitro. Coupled with the decreasing cost of sequencing, DNA has become a convenient readout for a variety of molecular biology assays. This started with the development of EST and cDNA technologies, was followed by high-throughput genome sequencing and then progressed through routine large-scale transcriptome sequencing, and finally to yet more intensive processes such as RNA-seq, Chip-seq and DNased-seq. We have even witnessed the development of DNA sequencing-based methods with no direct biological role, such as the mathematical exploration of a combinatoric space and the development of unique synthetic tags for property tracking.

DNA sequences determined for research purposes have been routinely archived since 1982, when the EMBL Data Library was founded. This was closely followed by the formation of GenBank first at the US Department of Energy and then transferred to NIH, and in 1987 by the DNA Databank of Japan. These three centres joined to form a tripartite collaboration, the INSDC, to archive and provide access to all DNA sequences generated by publicly funded research [3]. This data archiving project has gone through many changes in its 30-year history, responding both to advances in sequencing technology and to changes in the use of DNA sequence information.





Data Publishing







 $(GIGA)^n_{DB}$ 40 Datasets with DOI®s

Invertebrate

Ant

- Florida carpenter ant
- Jerdon's jumping ant
- Leaf-cutter ant

Roundworm

Schistosoma

Silkworm



Human

Asian individual (YH) v1+v2

- DNA Methylome
- Genome Assembly
- Transcriptome

Cancer (14TB)

Hep B infected exomes

Single Cell Bladder Cancer

Ancient DNA

- Saggaq Eskimo
- Aboriginal Australian

GENOME 10K.

Vertebrates

Giant panda

Macaque

- Chinese rhesus
- Crab-eating

Mini-Pig

Naked mole rat

Parrot

Penguin

- Emperor penguin
- Adelie penguin

Pigeon, domestic

Polar bear

Sheep

Tibetan antelope

Microbes

E. Coli O104:H4 TY-2482

Cell-Line

Chinese Hamster Ovary

Mouse Methylomes

Released pre-publication

Non-BGI

Paper in GigaScience

Plants

Chinese cabbage

Cucumber

Foxtail millet

Pigeonpea

Potato

Sorghum

Coming soon...

Microbiome data



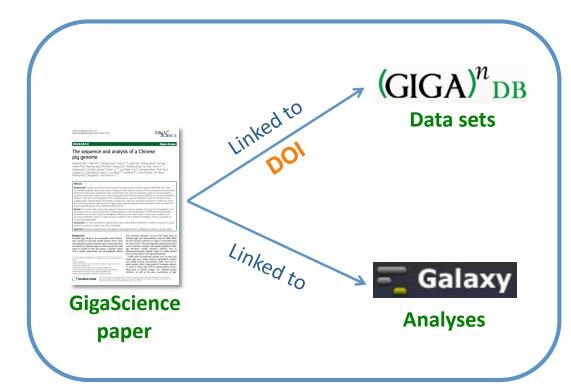


GigaDB v2 export to CBIIT GigaGalaxy

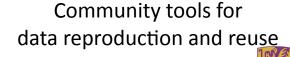
Results

| Species | Dataset type | Dataset | Sample | File type | File format | File name | Include in download |
|---|-----------------|--|---|--------------|----------------|---|------------------------|
| Human | Genomic | 10.5524/1000010 - Genomic sequence from an Aboriginal Australian | Biosample: 25 9765 - Aboriginal Australian human | SNPs | vcf 🕝 | AusAboriginal.hg19.var.fil tered.snps.sampled.vcf.gz | Ø |
| Human | Genomic | 10.5524/1000010 - Genomic sequence from an Aboriginal Australian | Biosample: 259765 - Aboriginal Australian human | SNPs | vcf 🚱 | AusAboriginal.hg19.var.fil tered.snps.vcf.gz | Ø |
| Link to GigaDB Link to sample if landing pages applicable | | Download to Galaxy Link to our documentation on file formats | | | | | |

How are we supporting data reproducibility?









CBIIT GigaGalaxy

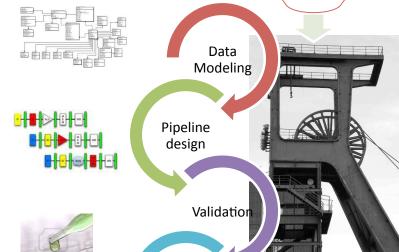
Data, Data, Data...



Galaxy







Applications





Tin-Lap Lee, CUHK





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- **GigaScience**
 - Scott Edmunds
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- myExperiment
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 - Dave De Roure



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Thank you



