

Synopsis about the Neurodevelopmental Bioinformatics Initiative (NBI)

Overview:

The mission of the NBI is to support the UAB community in the design and execution of experiments which utilize high-throughput sequencing technology. As one of the Civitan International Research Center's cores, the NBI is an interdisciplinary resource available to many researchers in campus across several departments.

Under the direction of Dr. Jeremy Day and Dr. Lara Ianov, the NBI provides services that fit project-specific requests (e.g.: publication ready figures, writing support in manuscripts) and large-scale requests (e.g.: pipeline execution) for comprehensive support of each study.

NBI model of services:

- **Next-generation sequencing analysis:** The long-term commitment of the NBI is to provide robust and reproducible pipelines for numerous omics applications (**Fig. 1**). Currently, the NBI provides support for several transcriptomics applications, such as bulk RNA-seq (whole-transcriptome, mRNA), single-cell/nuclei RNA-seq, CITE-seq and epigenomics applications such as whole-genome bisulfite sequencing (WGBS), reduced representation bisulfite sequencing (RRBS), and immunoprecipitation sequencing techniques such as MeDIP-seq, MBD-seq, ChIP-seq.
- **Visualization of omics data:** Presenting the wealth of information from an omics study is crucial for scientists to effectively relay their data. The NBI produces high-quality figures from omics data (**Fig. 1**).
- **Support for writing manuscripts and grant applications:** Formal assistance is provided during the writing/reviewing stages of manuscript and grants to ensure that the bioinformatics methods and data are properly described.
- **Bioinformatics training:** The NBI is happy to provide group or one-on-one training sessions to aid the community in the utilization of bioinformatics resources.

Representative workflow of service

GitLab

HPC

Interactive analysis & visualization

Pipeline configuration example

```
# sample list with basename of each fastq.gz
samples: "sk_supp/samples.txt"

# path to transcriptome reference
reference: "/data/user/lianov/Rattus_norvegicus.Rnor_6.0.cdna.all.fa"

# is the source from the transcriptome from GENCODE? 'yes' or 'no'
source_gencode: "no"

# path to GTF file
GTF: "/data/user/lianov/Rattus_norvegicus.Rnor_6.0.96.gtf"

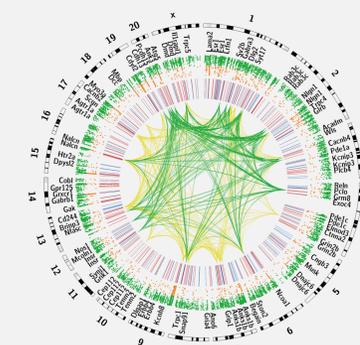
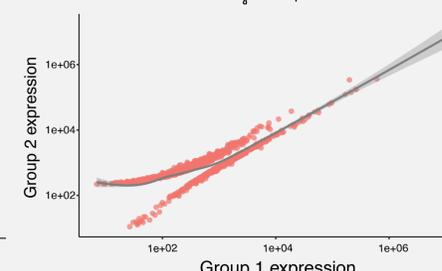
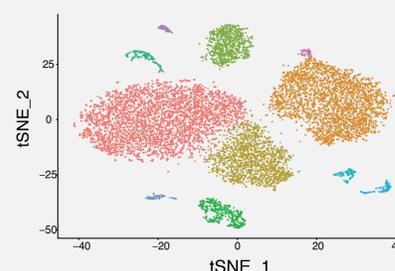
# Library type:
library_type: "ISR"
```

Pipelines powered by:

Snakemake
BIOCONDA

slurm
workload manager

R Studio
Bioconductor
OPEN SOURCE SOFTWARE FOR BIOINFORMATICS



Support options

Since the establishment of the NBI, we have provided free services to all of its users. However, changes in this model are needed to accommodate all user requests. Thus, effective **October 1st, 2019**, the following support options will be available (consultations and meetings will remain free):

- 1) **Effort from grants:** this option is best suited for users seeking long-term support for studies which will utilize next-generation sequencing. The effort incorporated should reflect the scope of the work.
- 2) **Standard service:** \$250 fixed rate + \$25 per hour for each project.
- 3) **Priority service:** \$1000 fixed rate + \$100 per hour for each project. This option provides the users with a choice of skipping the standard service queue to receive the results faster.

Contact information

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Figure 1: Illustration of a typical NBI workflow **1)** All code generated is maintained in GitLab, which serves as the starting point of any analysis where the NBI staff chose the appropriate pipeline to run and clones it to the High Performance Computer (HPC) cluster. **2)** All resource heavy analysis (e.g.: trimming, mapping, read quantification) are performed in HPC by setting up the pipeline configuration with the information relevant to the study in the parameters provided by the pipeline. The use of HPC, software environments (conda), and an advanced workflow management system (Snakemake), ensure that the analysis is performed in a fast, robust and reproducible manner. **3)** Once the analysis in HPC is complete, study-specific analysis (e.g.: statistical test which fits the user's experimental design) is performed to finalize the results. The NBI is also happy to assist any user with the visualization of omics data by providing publication ready figures. Custom requests of visualization are acceptable.