

# Building Infrastructure and Workflows for Clinical Bioinformatics

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**Citation:** Chong R (2021). Building Infrastructure and Workflows for Clinical Bioinformatics. *EJBI*. 17(4): 17

**DOI:** 10.24105/ejbi.2021.17.4.17

Received: March 27, 2021

Accepted: April 12, 2021

Published: April 19, 2021

## Editorial Note

Next generation sequencing (NGS) assays operate on large amounts of data, making bioinformatics an essential part of all NGS assays. These assays have been advancing both in size, from small gene panels to clinical whole genome sequencing, as well as complexity of examination, and number and types of analytes.

Clinical molecular laboratories executing NGS testing must be in conformity with accreditation checklist requirements provided by the College of American Pathologists (CAP) and regulations listed by the clinical Laboratory Improvement Revisions. The Association for Molecular Pathology (AMP) and CAP provided suggestions for validation of clinical bioinformatics pipelines including their design and development. However, factors about computational infrastructure and pipeline frameworks are few across all these recommendations.

One of the main purposes for this is that there is lack of regularization in implementation of bioinformatics pipelines for clinical NGS assays and there are multiple reasons for this. The range of assays obtainable in each laboratory is vast, and these assays differ in technology, design, target analytes, and certain restrictions of detection. Even if the same assay is implemented at various institutions, there is a lot of irregularity due to major differences in hardware and bioinformatics pipeline orchestration. Computing paradigms and technology vary extremely across institutions, which makes flexibility and standardization very difficult. The availability of skilled bioinformatics staff also can contribute to these differences.

A laboratory might not have the capability to design complex pipeline modules in the absence of accomplished staff and might adopt less-flexible out-of-the-box vendor contributed solutions. Processing and examination of genomic data is an example of a big data application requiring complex interdependent data examination workflows. Such bioinformatics assignments take the primary and secondary input data through several computationally-intensive transforming steps using variable software packages, where some of the outputs form inputs for other steps. Implementing scalable, reproducible, portable and easy-to-use workflows is particularly demanding.

The interdisciplinary nature of bioinformatics and genomics data examination calls for a bioinformatics pipeline that advances collaboration and reflects the way you can most efficiently and dependably process and analyze genomic data – now and into the future. Setting up a new next-generation sequencing (NGS) pipeline or simply adding components to an existing pipeline needs careful thought and planning.

## Conclusion

A laboratory might not have the capability to design complex pipeline modules in the absence of accomplished staff and might adopt less-flexible out-of-the-box vendor contributed solutions. One of the main purposes for this is that there is lack of regularization in implementation of bioinformatics.