From Fastq To Drug Recommendation – Automated Cancer Report Generation using OncoRep & Omics Pipe

Tobias Meissner[1,3*], Kathleen M. Fisch[2,3*], Louis Gioia[3] & Andrew I. Su[3]

[1] Department of Molecular and Experimental Medicine, Avera Cancer Institute, La Jolla, CA, USA.
Email: tobias.meissner@avera.org

[2]Department of Medicine, Center for Computational Biology & Bioinformatics, University of California, San Diego, La Jolla, CA, USA.

[3] Department of Molecular and Experimental Medicine, The Scripps Research Institute, La Jolla, CA, USA.

* both authors contributed equally to this work

Project Website: http://sulab.org/tools/oncorep-oncogenomics-report/
Source Code: https://bitbucket.org/sulab/oncorep/ & https://bitbucket.org/sulab/omics_pipe/

Abstract

Next generation sequencing allows us to study cancer in a large scale and in multiple dimensions. This provides in-depth insight into tumor pathogenesis on the individual patient level, paving the path to precision medicine. The advent of precision medicine introduces a shift in how cancer patients will be treated in the future, away from the one drug-one disease paradigm towards the idea of bringing the right drug to the right patient.

Challenges that arise with this paradigm shift are i) High-throughput, automated, parallel, and reproducible processing and analysis of sequencing data in the n-of-1 setting, ii) extracting, integrating, interpreting and reporting relevant information from various layers of omics data iii) integrating omics data with drug databases, (iv) presenting the information in an understandable and timely manner to provide clinically relevant and actionable targets to the clinician and tumor board, and (v) creating a transparent, open-source, community framework for enabling community curation of best practices and algorithms, and reproducible analysis pipelines in a move towards standardizing next generation sequencing analysis for patient care.

To address these challenges, we present an automated cancer reporting framework based on two open-source platforms, OncoRep and Omics Pipe. OncoRep is an RNAseq-based n-of-1 reporting tool for cancer patients that has been integrated into Omics Pipe, a community-based framework that enables reproducibility by curating and automating best practice omics data analysis pipelines. We have applied this framework to breast cancer patients in an n-of-1 setting. RNA-seq was performed for each patient and the fastq files were processed and analyzed using Omics Pipe, generating expression data, variants and fusions based on published methods. OncoRep was incorporated as a custom pipeline in Omics Pipe to perform prospective molecular classification, detect altered genes and pathways, identify gene fusion events and clinically actionable mutations, and to report suitable drugs based on identified actionable targets. Omics Pipe automates these analyses and keeps a detailed record of the analyses and parameters used to ensure reproducibility. OncoRep integrates and visualizes the data in an approachable html based interactive report as well as a PDF based summary report, providing the clinician and tumor board with an approachable report to guide the treatment decision making process. We hope with our contribution to enhance transparency and reproducibility of next generation sequencing analysis and clinical reporting, which will be needed for moving this technology into routine clinical practice.