

Terra: Your cloud-native analysis platform

ASHG 2020



Terra as Orchestrator



"[Terra is] a platform that allows bioinformatics researchers to take advantage of the cloud's power to scale without having to deal with the underlying complexity."

- [Genomics in the Cloud](#) (Brian D. O'Connor and Geraldine Van der Auwera)



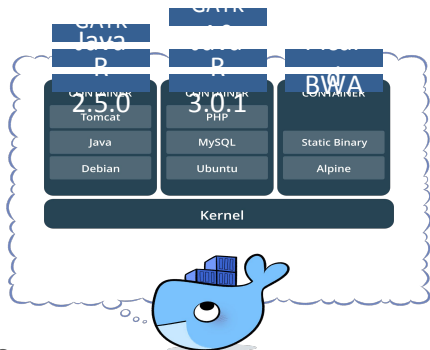
Sets up and manages resources

- Cloud-based compute and storage
- Data access and security

Runs analysis

Stores generated data

Sets up the analysis environment - in a reproducible, shareable way



The Workspace - fundamental unit in Terra



Navigation bar for Terra Workspaces. It includes a menu icon, the Terra logo, 'POWERED BY Terra WORKSPACES', a 'BETA' badge, the text 'Workspaces > help-gatk/Reproducibility_Case_Study_Tetralogy_of_Fallot (read only)', a 'Notebook Runtime STOPPED (\$0.05 hr)' indicator, and a settings gear icon. Below this is a secondary navigation bar with 'DASHBOARD', 'DATA', 'NOTEBOOKS', 'WORKFLOWS', and 'JOB HISTORY' tabs, and a help icon.

ABOUT THE WORKSPACE

Reproducing the paper: Variant analysis of Tetralogy of Fallot

Overview

This workspace reproduces the work described by Matthieu Miossec and collaborators in the bioRxiv preprint "[Deleterious genetic variants in NOTCH1 are a major contributor to the incidence of non-syndromic Tetralogy of Fallot \(ToF\).](#)"

The original ToF study is a classic example of a study to understand the genetics that underlie a particular phenotype. The workspace reproduces all steps in the study as closely as possible, from processing the raw data (BAM) files, to calling variants, to the clustering analysis that led to the final result.

The workspace serves as a template of best practices for making your own work easily reproducible with a detailed explanation of how we reproduced the ToF study using a cloud-based analysis platform. Sample data and notebooks allow users to reproduce the process themselves.

Summary of original ToF study

By analysing high-throughput exome sequence data from 867 cases and 1252 controls, the authors identified 49 deleterious variants within the NOTCH1 gene that appeared to correspond with the ToF congenital heart disease. Others had previously identified NOTCH1 variants in families with congenital heart defects, including ToF. However, the work by Miossec et al. is the first to scale variant analysis of ToF to a cohort of nearly a thousand case samples and show that NOTCH1 is a significant contributor to ToF risk.

- **Paper URL:** <https://www.ahajournals.org/doi/abs/10.1161/CIRCRESAHA.118.313250> (This workspace is based on the [preprint](#))
- **Workflows:** see [Workflows](#) tab
- **Notebook:** [download from Google Cloud Storage](#)
- **Precomputed results:** [view in Google Cloud Storage](#)

WORKSPACE INFORMATION

CREATION DATE 3/15/2019	LAST UPDATED 5/12/2020
SUBMISSIONS 0	ACCESS LEVEL Reader

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TAGS

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