



The Elizabeth H.
and James S. McDonnell III

**MCDONNELL
GENOME INSTITUTE**

at Washington University



Washington
University in St. Louis

SCHOOL OF MEDICINE

PMBIO Module 04

Germline. Germline WGS and Exome Variant Analysis

Malachi Griffith, Obi Griffith, Zachary Skidmore, Huiming Xia
Introduction to bioinformatics for DNA and RNA sequence
analysis (IBDR01)

29 October - 2 November, 2018

Glasgow



Attribution-ShareAlike 4.0 International (CC BY-SA 4.0)

This is a human-readable summary of (and not a substitute for) the [license](#). [Disclaimer](#).

You are free to:

Share — copy and redistribute the material in any medium or format

Adapt — remix, transform, and build upon the material for any purpose, even commercially.

The licensor cannot revoke these freedoms as long as you follow the license terms.



Under the following terms:



Attribution — You must give [appropriate credit](#), provide a link to the license, and [indicate if changes were made](#). You may do so in any reasonable manner, but not in any way that suggests the licensor endorses you or your use.



ShareAlike — If you remix, transform, or build upon the material, you must distribute your contributions under the [same license](#) as the original.

No additional restrictions — You may not apply legal terms or [technological measures](#) that legally restrict others from doing anything the license permits.

Learning objectives of module 04: Germline

- **Key concepts:** Variation, germline mutation, polymorphism, SNPs, variant databases, germline variant calling
- Use of the GATK tool kit for variant calling
- Perform basic germline variant filtering
- Compare single sample germline variant calling to cohort based calling