



Cold
Spring
Harbor
Laboratory

Advanced Sequencing Technologies & Applications

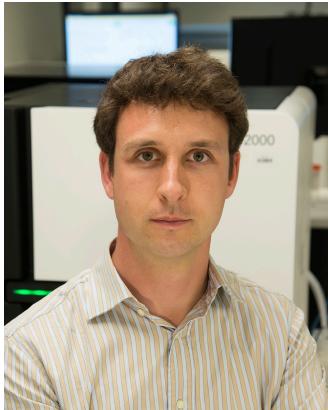
<http://meetings.cshl.edu/courses.html>

Introductions to MGI/WashU instructors



Malachi Griffith

Assistant Professor of Medicine
Assistant Professor of Genetics
Assistant Director, MGI



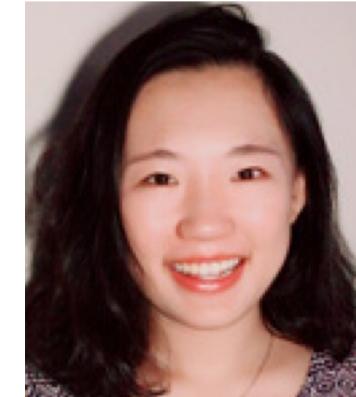
Obi Griffith, PhD

Associate Professor of Medicine
Assistant Professor of Genetics
Assistant Director, MGI



Felicia Gomez

Instructor of Medicine



Huiming Xia

PhD candidate



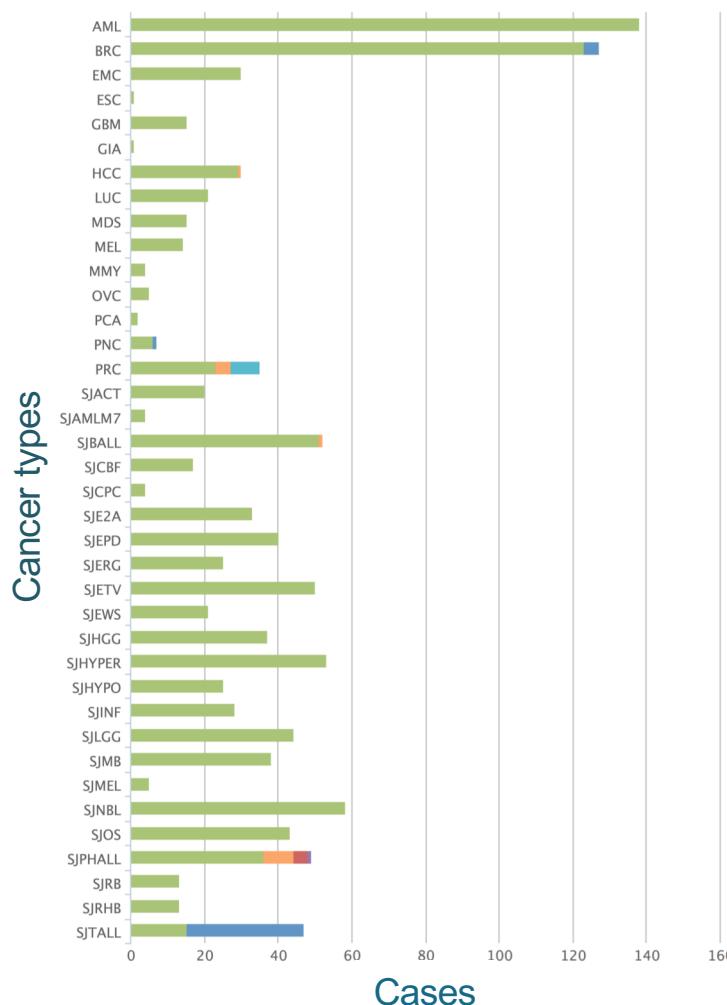
www.griffithlab.org



Kelsy Cotto
PhD candidate

McDonnell Genome Institute, Washington University School of Medicine

Griffith Lab activities are cancer genomics (informatics) focused: new discovery, translation, method development and training



- **Cancer genome analysis**
 - Lymphoma
 - Breast cancer
 - Mouse models
 - Targeted sequencing of ER+ cohorts
 - Non-coding mutation analyses
 - Small cell lung cancer
 - OSCC
 - ...
- **NGS Technology**
 - Regulome sequencing
 - TCR repertoire sequencing
- **Precision Medicine for Cancer**
 - [Genomics Tumor Board](#)
 - [Immunotherapy](#) (cancer vaccines)
- **Tool development**
 - [RegTools](#) – Regulatory/splicing variant analysis
 - [genVisR](#) – Genome Visualizations in R
 - [DGIdb](#) – drug-gene interactions database
 - [CIViC](#) – Clinical Interpretation of Variants
 - [pVACtools](#) – Cancer vaccine designs
 - [VAtools](#) – VCF annotation
- **Bioinformatics Training**
 - RNA-seq analysis ([rnabio.org](#))
 - Genomic Data Visualization ([genviz.org](#))
 - Precision Medicine Bioinformatics ([pmbio.org](#))
 - [CSHL](#) and [CBW](#)

www.griffithlab.org

Introduction to SEQTEC Informatics – philosophy and goals

Do “the bioinformatics” for someone, and you help them for a day. Teach someone to do bioinformatics, and you help them for a lifetime.

- Ancient Chinese proverb

- Course goals
 - Learn concepts and develop skills for sequence analysis
 - Build the foundation for tackling your own analysis challenges
 - Learn to think like a bioinformatician
 - Have fun

Outline for informatics portion of course

Day 1: Unix, Cloud Computing, DNA sequence alignment

Day 2: RNAseq - installation, data/files, alignment, QC

Day 3: RNAseq – count-based data, expression, DE

Day 4: scRNA – data, QC, visualization, expression, ATAC-seq

Day 5: scRNA – exercises; variant discovery; **Banquet**

Day 6: Variant analysis and interpretation

Day 7: Genome arithmetic (BEDtools)

Day 8: Long read sequence analysis

Day 9: Pipelines, scripting, wrap up

Course format for a typical day

- [open Q&A and discussion]
- Lecture
- BREAK
- Lunch
- Practical exercises
- BREAK
- Practical exercises
- Dinner
- practical exercises
- Team assignments

Informatics background poll

- Student poll
 - Are you doing genomics research?
 - Do you consider yourself a bioinformatician?
 - What kind of NGS data are you working with?
 - WGS?
 - Exome?
 - RNAseq?
 - Epigenome?
 - Single cell?
 - Other?
 - What organism do you work with?
 - Did you bring data?

Student poll continued

Not counting the pre-requisites and materials for this course:

- Are you familiar with linux/command line?
 - Intermediate?
 - Expert?
- Do you sometimes write code?
 - What language?
- Are you familiar with R?
 - Intermediate?
 - Expert?
- Do you use git/github?

An overview of bioinformatics

Adam Siepel

Professor, Watson School of Biological Sciences; Chair, Simons Center for Quantitative Biology; Cold Spring Harbor Laboratory

Opinion | [Open Access](#) | Published: 29 July 2019

Challenges in funding and developing genomic software: roots and remedies

[Adam Siepel](#) 

[Genome Biology](#) 20, Article number: 147 (2019) | [Cite this article](#)

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