



Cold
Spring
Harbor
Laboratory

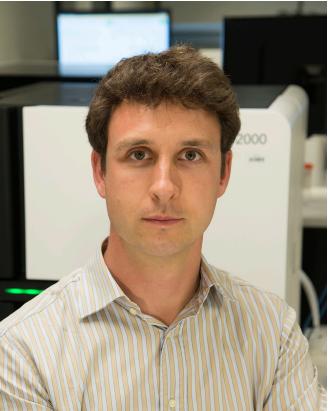
Advanced Sequencing Technologies & Applications

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

Introductions to MGI/WashU instructors



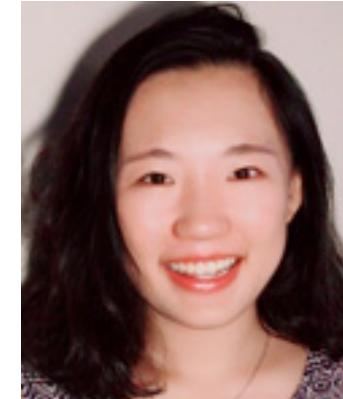
Malachi Griffith
Assistant Professor of Medicine
Assistant Professor of Genetics
Assistant Director, MGI



Obi Griffith, PhD
Associate Professor of Medicine
Assistant Professor of Genetics
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Felicia Gomez
Instructor of Medicine



Huiming Xia
PhD candidate



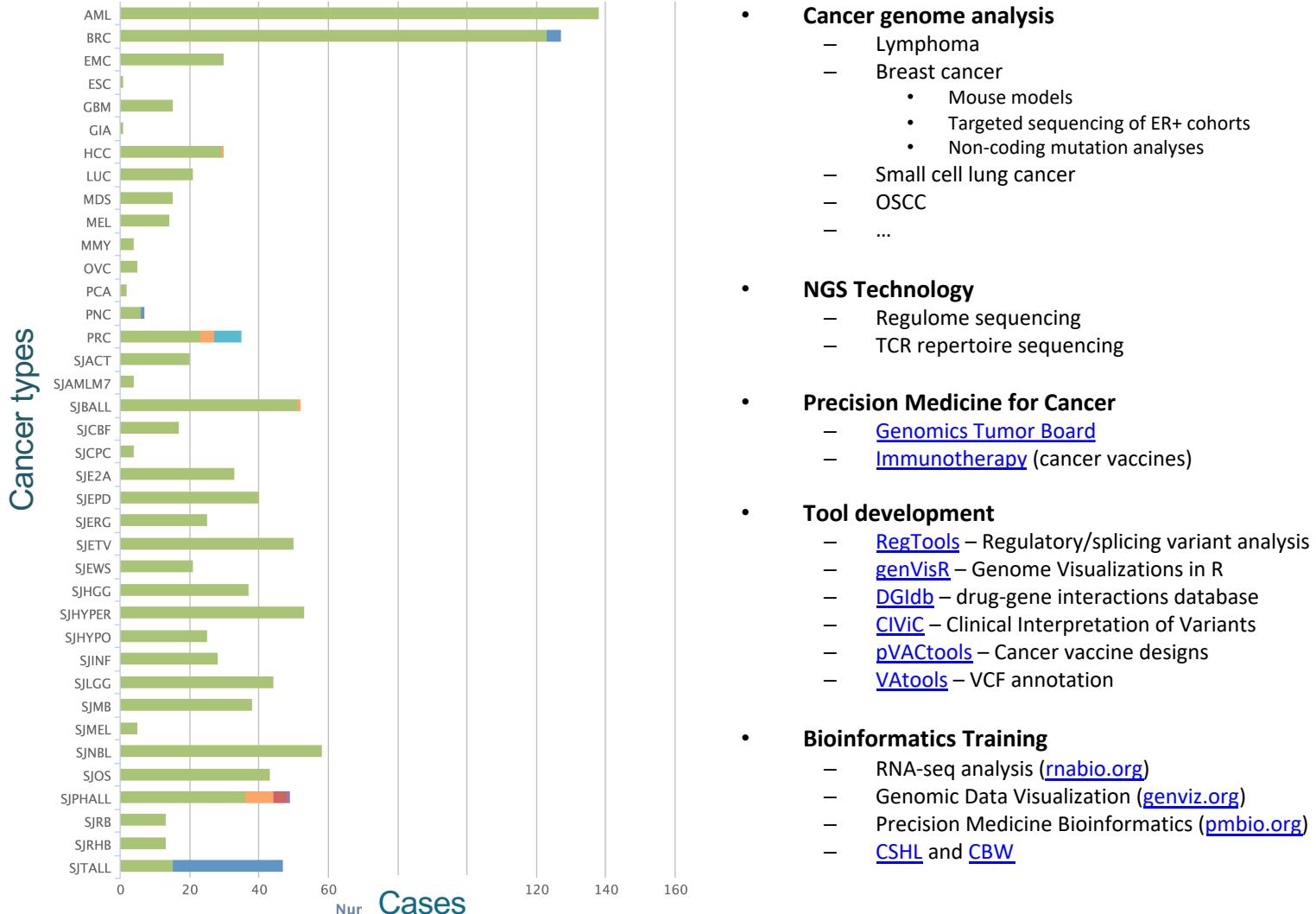
www.griffithlab.org



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](#)

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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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