**Cold Spring Harbor Advanced Sequencing Technologies and Applications**
**Mon Nov 5 - Sun Nov 18, 2018 (Course days: November 6 - 18, 2018)**

**Prerequisites**
These are recorded in the CSHL Wiki (Link). Students were provided with this list in advance of the course by Alicia Franco (afranco@cshl.edu).

**Mealtimes at Blackford Hall**
Breakfast: 7:30am - 9:00am
Lunch: 11:30am - 1:30pm
Dinner: 5:30pm - 7:00pm

**5th November (Monday)**
7:00pm: Meet and greet with instructors. In the Blackford Hall Bar (lower level) for introductions and course overview

**6th November (Tuesday)**
9:00am - 9:30am: Introduction to the course and curriculum. Safety presentation. Review of activities, group assignments, schedule. Q&A.
9:30am - 10:30am: Overview of Next-generation Sequencing Technologies (Elaine Mardis)
10:30am - 11:00am: **BREAK**
11:00am - 12:00pm: DNA Fragmentation and QC for NGS Library Prep
12:00pm - 1:00pm: **LUNCH**
1:00pm - 2:00pm: Library Prep methods (WGS, 10X Genomics, PacBio, Tagmentation, UMIs)
2:00pm - 3:30pm: Target Enrichment (Hybrid Capture)
3:30pm - 4:30pm: RNA-Seq Processes (Prep, QC, Library approaches)
4:30pm - 5:30pm: “Ethics in Genetics and Genomics Research” (Jan Witkowski)
5:30pm - 7:00pm: **DINNER**
7:00pm - 9:00pm: 10 Student presentations (5-7 minute talk focusing upon your research and planned use of NGS)
9:00pm - 10:00pm: **SOCIAL HOUR (after student talks)**

**7th November (Wednesday)**
9:00am - 10:30am: Aravinda Chakravarti lecture. "Genetic Regulatory Control of Cardiac Diseases"
10:30am - 10:45am: **BREAK**
10:45am - 12:00pm: Introduction to Long read sequencing (Sara Goodwin)
12:00pm - 12:30pm: Classroom: Overview of ONT lab section (Sara Goodwin)
12:30pm - 1:30pm: **LUNCH**
1:30pm - 2:30pm: First strand synthesis, cDNA PCR, Ligation, bead binding (Sara Goodwin)
2:30pm - 3:00pm: Travel to Woodbury (by shuttle, confirmed by Alicia Franco)
3:00pm - 3:30pm: Prepare and load GridION (Sara Goodwin)
3:30pm - 4:30pm: Tour of Woodbury Genome Center
4:30pm - 5:00pm: Travel Back to CSHL
5:30pm - 6:30pm: **DINNER**

6:30pm - 9:00pm: 10 Student presentations (5-7 minute talk focusing upon your research and planned use of NGS)
9:00pm - 10:00pm: **SOCIAL HOUR (after student talks)**

**8th November (Thursday)**
9:00am - 10:15am: "Dissecting the RNA-interactome with NGS" (Chris Maher)
10:15am - 10:30am: **BREAK**
10:30am - 11:30am: Single Cell Transcriptomics lab section: begin tissue dissociation protocol - set up ~2 hour incubation
11:30am - 12:30pm: “Single cell sequencing technology and applications” (Jonathan Preall)
12:30pm - 1:30pm: **LUNCH**
2:00pm - 5:00pm: Finish tissue dissociation, column-enrich live cells, perform 10X cell capture and begin single cell RT-PCR. (T.A. will finish rest of library prep offline)
5:00pm - 5:30pm: **BREAK**
5:30pm - 7:00pm: **DINNER (Wine and cheese followed by pizza)**

**9th November (Friday)**
9:00am - 10:30am: **MORNING OFF**
10:30am - 12:00pm: Introduction to Cloud Computing (Kelsy Cotto)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 2:30pm: Introduction to NGS data analysis lecture (Sorana Morrissy)
2:30pm - 2:45pm: **BREAK**
2:45pm - 4:00pm: “Identifying Driver Alterations and Therapeutic Options in Cancer” (Debyani Chakravarty)
5:30pm - 6:30pm: **DINNER**
6:30pm - 8:00pm: Introduction to Unix commands and file system (Alex Wagner)
8:00pm - 8:15pm: **BREAK**
8:15pm - 9:30pm: Introduction to Unix commands and file system continued (Alex Wagner)

**10th November (Saturday)**
9:00am - 10:25am: FASTQ format & sequence alignment overview (Andrew Farrell)
10:25am - 10:35am: **BREAK**
10:35am - 12:00pm: BAM format and samtools tutorial (Alistair Ward)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 3:30pm: IGV tutorial (Kelsy Cotto)
3:30pm - 4:00pm **BREAK**
4:00pm - 6:00pm: Annotations, UCSC/Ensembl, BED/GFF, BigWig, Tabix (Alistair Ward)
6:00pm - 7:00pm: **DINNER**
**7:00pm - 8:00pm: Open Q&A and discussion**
8:00pm - 11:00pm: **EVENING OFF**

**11th November (Sunday)**
8:30am - 9:30am: Variant discovery lecture (Andrew)
9:30am - 9:45am: **BREAK**
9:45am - 12:00pm: Variant discovery practical (Andrew Farrell)

12:00pm - 1:00pm: **LUNCH**
1:00pm - 3:00pm: Variant discovery, VCF format, and practical exercise (cont.) (Andrew Farrell)
3:00pm - 3:30pm: **BREAK**
3:30pm - 5:30pm: Variant annotation, prioritization and visualization (IOBIO) (Alistair Ward)
5:30pm - 7:00pm: **DINNER**
**7:00pm - 8:00pm: Open Q&A and discussion**
8:00pm - 11:00pm: **EVENING OFF**

**12th November (Monday)**
9:30am - 12:00pm: Disease variant discovery (Aaron Quinlan)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 3:30pm: Disease variant discovery session with GEMINI (practical session, disease gene discovery) (Aaron Quinlan)
3:30pm - 5:00pm: RNA-seq, Intro to RNA-sequencing lecture (Malachi Griffith)
5:00pm - 6:00pm: RNA-seq, Intro to RNA-sequencing lab (Obi Griffith)
6:00pm - 7:00pm: **DINNER**
7:00pm - 7:30pm: RNA-seq, Alignment and visualization lecture (Obi Griffith)
7:30pm - 9:00pm: RNA-seq, Alignment and visualization lab (Malachi Griffith)

**13th November (Tuesday)**
9:00am - 9:30am: RNA-seq, Expression and differential expression lecture (Jason Walker)
9:30am - 11:00am: RNA-seq, Expression and differential expression analysis lab (Obi Griffith)
11:00am - 11:15am: **BREAK**
11:15am - 12:30pm: RNA-seq, Differential expression visualization lab (Jason Walker)
12:30pm - 1:30pm: **LUNCH**
1:30pm - 3:45pm: RNA-seq, Reference-free expression analysis lab (Malachi Griffith)
3:45pm - 4:00pm: **BREAK**
4:00pm - 5:00pm: Monoallelic expression of human genes in diverse tissues (Stephanie Kravitz)
5:00pm - 6:00pm: **FREE TIME TO RELAX AND PREPARE FOR BANQUET**
6:00pm - 8:00pm: **BANQUET DINNER - Confirmed with Rachel Lopez on 8/28/18 - WRM**
8:00pm - 11:00pm: **EVENING OFF**

**14th November (Wednesday)**
9:00am - 10:00am: Transcript assembly lecture (Brian Haas)
10:00am - 10:15am: **BREAK**
10:15am - 12:00pm: Transcript assembly lab (Brian Haas)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 3:00pm: Gene regulation, allelic expression, QC from GTeX (Tuuli Lappalainen)
3:00pm - 3:15pm: **BREAK**
3:15pm - 4:30pm: Joe Pickrell lecture. “Variant calling and disease risk prediction from low-pass sequencing data”.
**4:30pm - 5:30pm: Open Q&A and discussion**
5:30pm - 6:30pm: **DINNER**
6:30pm - 9:30pm: Set up (Ken Dewar)

**15th November (Thursday)**
8:30am - 9:30am: PacBio Introduction and setup (Ken Dewar)
9:30am - 9:45am: **BREAK**
9:45am - 12:00pm: Intro to Genome Assembly of PacBio + Illumina data (Ken Dewar)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 3:00pm: Assembly and Annotation (Ken Dewar)
3:00pm - 3:15pm: **BREAK**
3:15pm - 6:00pm: Assembly and Annotation cont'd (Ken Dewar)
6:00pm - 7:00pm: **DINNER**
7:00pm - 10:00pm: Annotation and analysis (Ken Dewar)

**16th November (Friday)**
9:00am - 10:00am: "Functional interpretation of non-coding sequence variants" lecture (Ekta Khurana)
10:00am - 10:15am: **BREAK**
10:15am - 12:00pm: Functional interpretation of non-coding sequence variants workshop (Ekta Khurana)
12:00pm - 1:00pm: **LUNCH**
1:00pm - 2:30pm: Genome Arithmetic with BEDTOOLS (Aaron Quinlan)
2:30pm - 3:00pm **BREAK**
3:00pm - 6:00pm BEDTOOLS tutorial and challenge problems (Aaron Quinlan)
6:00pm - 8:00pm: **DINNER (Pizza at Hershey)** and open discussion of specific topics
8:00pm - 11:00pm: **EVENING OFF**

**17th November (Saturday)**
9:00am - 10:00am: Review student's 10X Illumina Single Cell RNA-seq results (Jonathan Preall)
10:00am - 10:15am **BREAK**
10:15am - 12:30pm: Structural variation lecture and practical (Aaron Quinlan)
12:30pm - 1:30pm: **LUNCH**
1:30pm - 3:30pm: Probability and statistics for genomics analysis (Aaron Quinlan)
3:30pm - 4:00pm: Wrap up and Evaluations
4:00pm - 6:00pm: **BREAK**
6:00pm - 7:00pm: **DINNER**
7:00pm - 11:00pm: **EVENING OFF or TRAVEL**

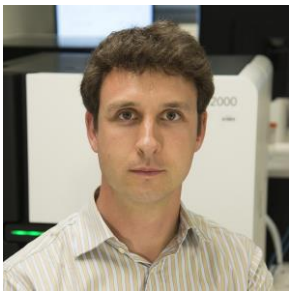
**18th November (Sunday)**
**TRAVEL DAY**

**Student list**

| | | |
|---|---|---|
| Dr. Karen A Cadoo | NY | cadook@mskcc.org |
| Dr. Maria I Carlo | NY | carlom@mskcc.org |
| Dr. Paul A Donat | NY | paul.donat@stonybrook.edu |
| Dr. Gloria R Garcia | MD | gloria.garcia@nih.gov |
| Dr. Maria G Gervasi | MA | mariagracia@vasci.umass.edu |
| Ms. Nathalia Graf Grachet | OK | ngrafgr@okstate.edu |
| Dr. Vikas A Gupta | GA | vikas.gupta@emory.edu |
| Dr. Peter J Kilfeather | United Kingdom | peter.kilfeather@dpag.ox.ac.uk |
| Mr. Seung J Kim | Canada | skim823@uwo.ca |
| Mr. Tomohiro Kumon | PA | kumon@sas.upenn.edu |
| Ms. Dawn M Layman | NY | dlayman@estee.com |
| Ms. Christine S Liu | CA | csl022@ucsd.edu |
| Mr. Ahmed M Malik | MI | ahmalik@umich.edu |
| Ms. Shaimaa F. Mouftah Ali | United Arab Emirates | 201590053@uaeu.ac.ae |
| Mr. Diego Rodriguez Terrones | Germany | diego.rodriguez@helmholtz-muenchen.de |
| Dr. Alec Sexton | CT | alec.sexton@yale.edu |
| Dr. Zonggao Shi | IN | zshi2@nd.edu |
| Dr. Francesco R Simonetti | MD | fsimonetti@jhmi.edu |
| Mr. Tianhao Xu | IL | tianhao.xu@my.rfums.org |
| Ms. Qinyu Sun | IL | qsun9@illinois.edu |
| Ms. Xiaoli Wu | NY | xlw1207@gmail.com |

**Photos: Course Instructors**

Obi Griffith, WASHU



Malachi Griffith, WASHU

Elaine Mardis, NCH

W. Richard McCombie, CSHL

Aaron Quinlan, UU

**Photos: Other Instructors, TAs, and lecturers**

Jon Belyeu, UU

Aravinda Chakravarti, NYU

Debyani Chakravarty, MSK

Kelsy Cotto, WASHU

Ken Dewar, McGill

Matt D'Iorio, McGill

Andrew Farrell, UU

Sara Goodwin, CSHL

Brian Haas, Broad

Stephanie Kravitz, UU

Ekta Khurana, Cornell

Tuuli Lappalainen, Columbia

Christopher Maher, WASHU

Sorana Morrissy, UC

Joe Pickrell, Gencove

Jonathan Preall, CSHL

Tuan Trieu, Cornell

Jason Walker, WASHU

Alex Wagner, WASHU

Robert Wappel, CSHL

Alistair Ward, UU

Jan Witkowski, CSHL