Genomic Resources

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QB Bootcamp Lecture 3

Outline

Part 1: Overview & Fundamentals

Part 2: Sequence Analysis Theory

Part 3: Genome Resources

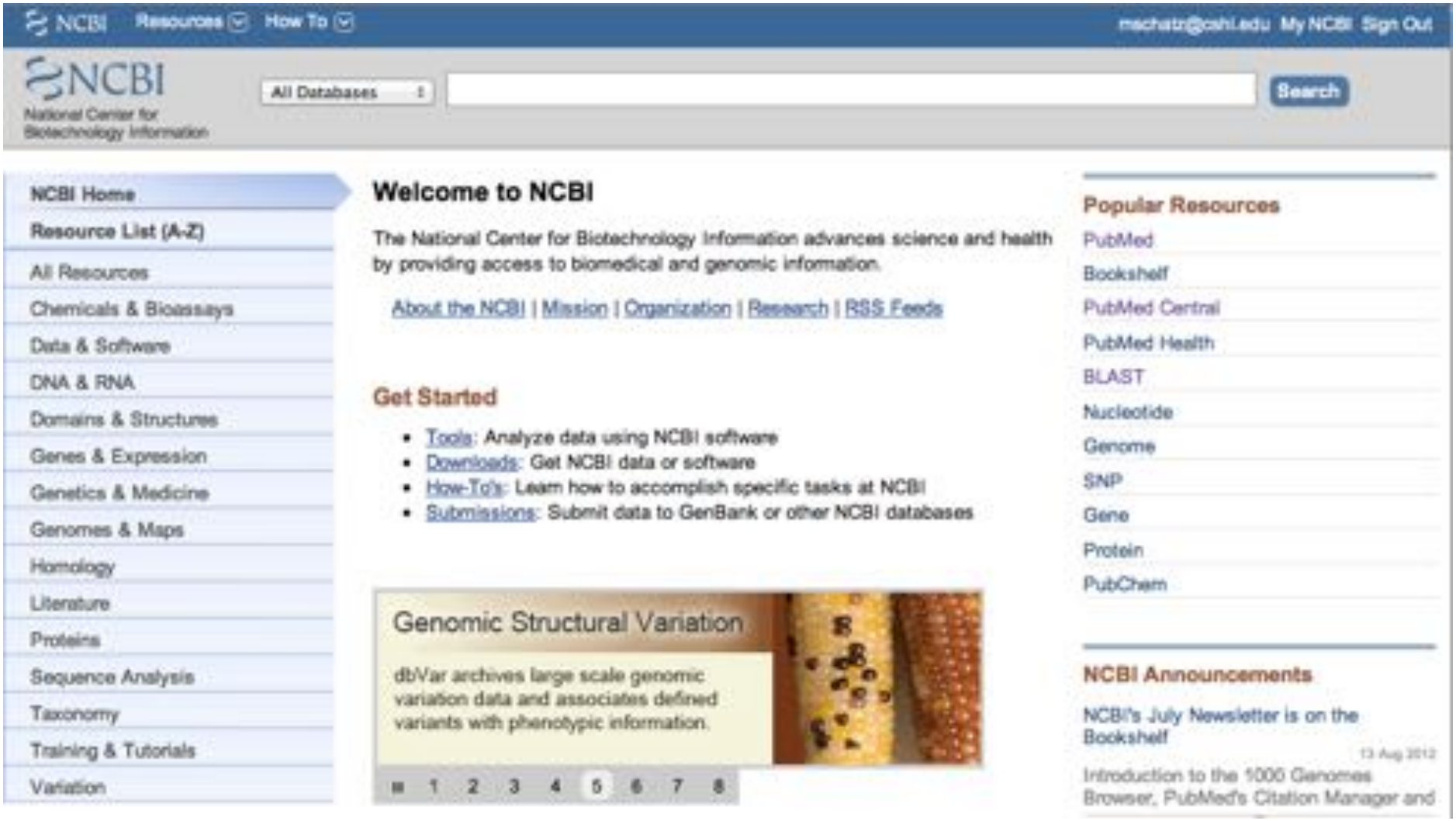
- Public: NCBI, UCSC
- CSHL: Intranet, Meetings, Galaxy

Part 4: Unix Scripting

Part 5: Example Analysis

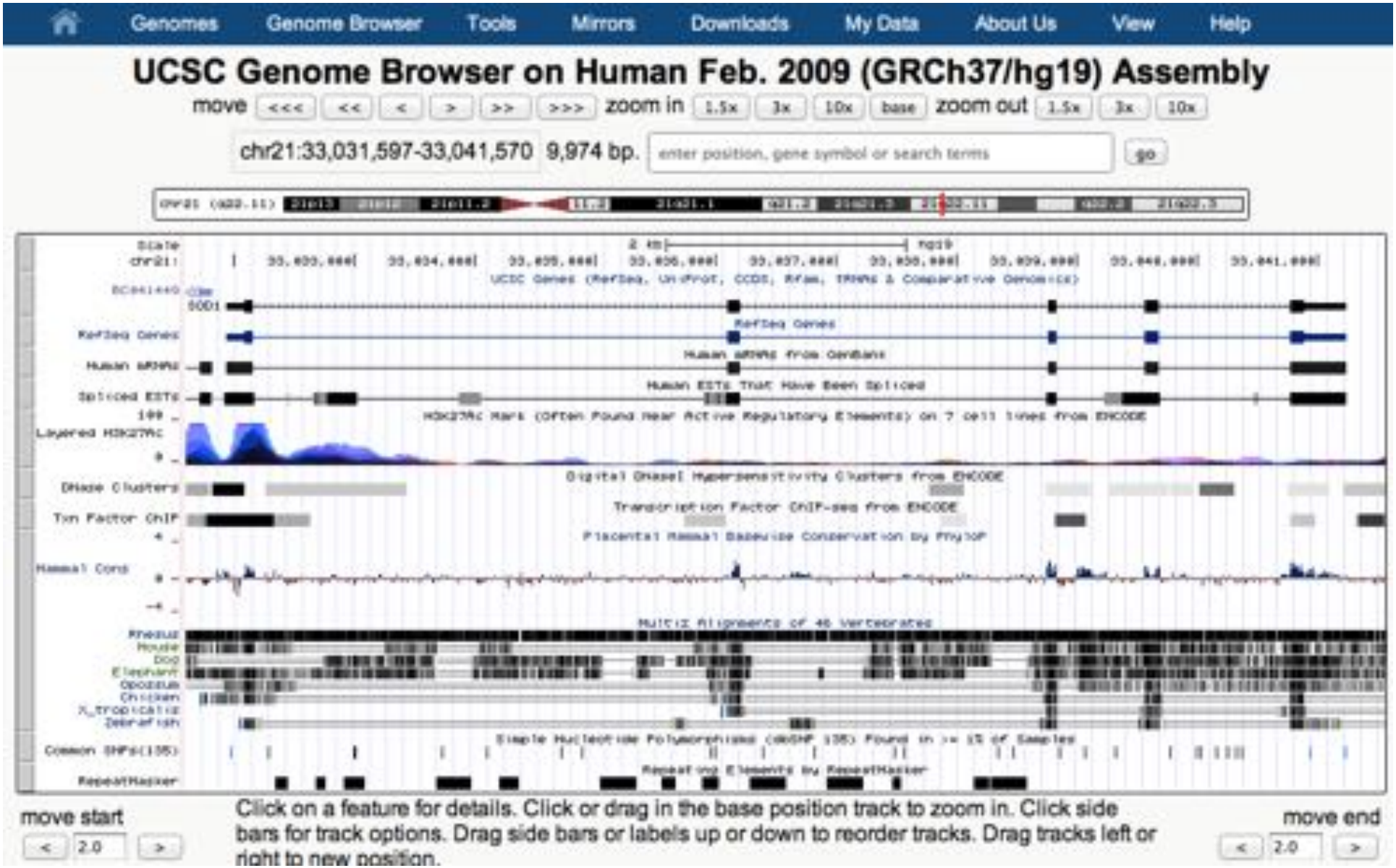
NCBI

http://www.ncbi.nlm.nih.gov/



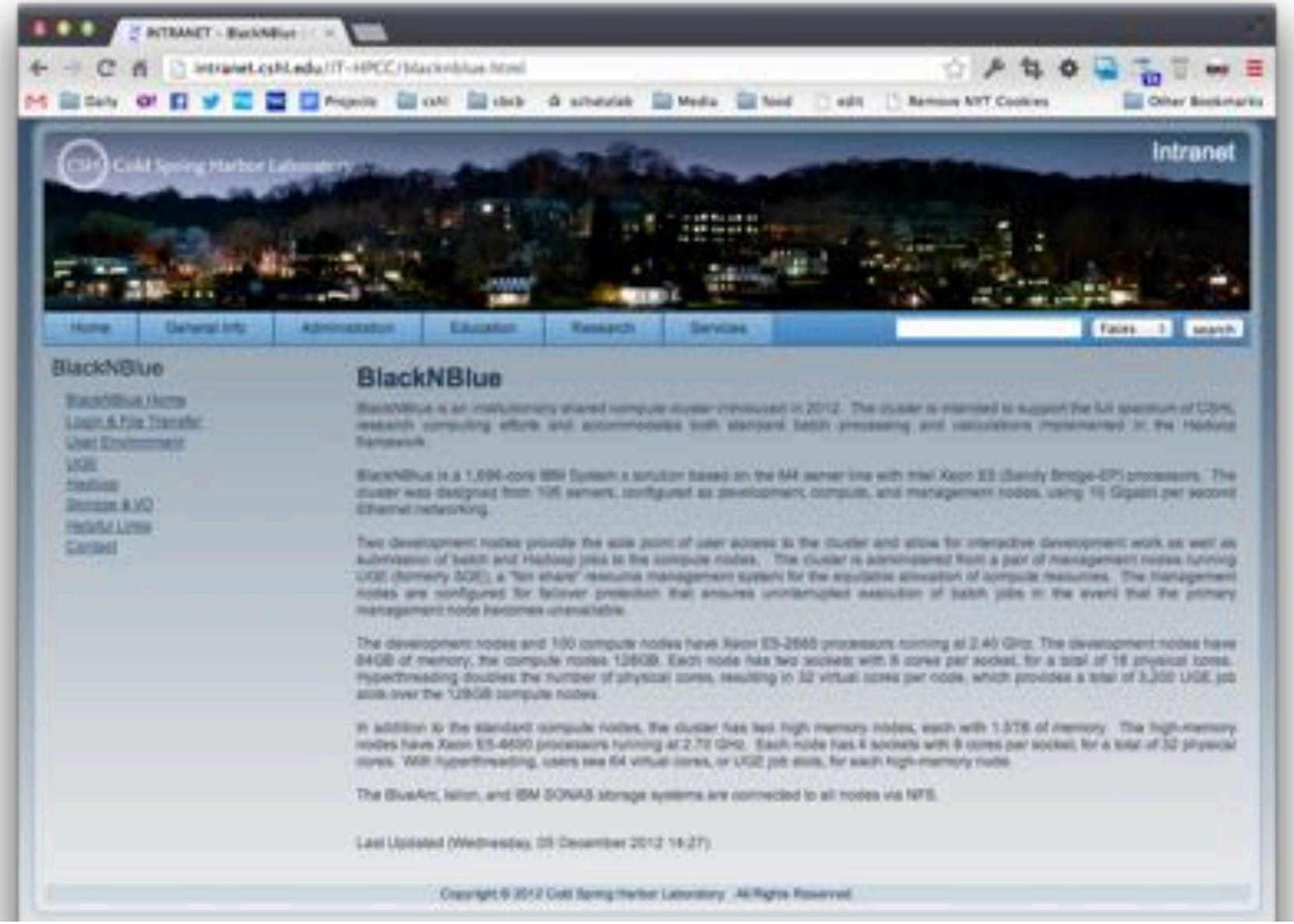
UCSC Genome Browser

http://genome.ucsc.edu/



Intranet

http://intranet.cshl.edu/IT-HPCC/blacknblue.html



Conferences and Journals

CSHL Yearly Conferences

Biology of Genomes	May	Latest advances in biology, genomics, and medicine
Symposium	May/June	Latest advances with yearly themes
Genome Informatics	Sept/Nov	Computational Biology
Personal Genomes	Sept/Nov	Computational Biology
In-house Symposium	Nov	Updates from the faculty (Just before Thanksgiving)

You are welcome to attend all meetings at CSHL free of charge:

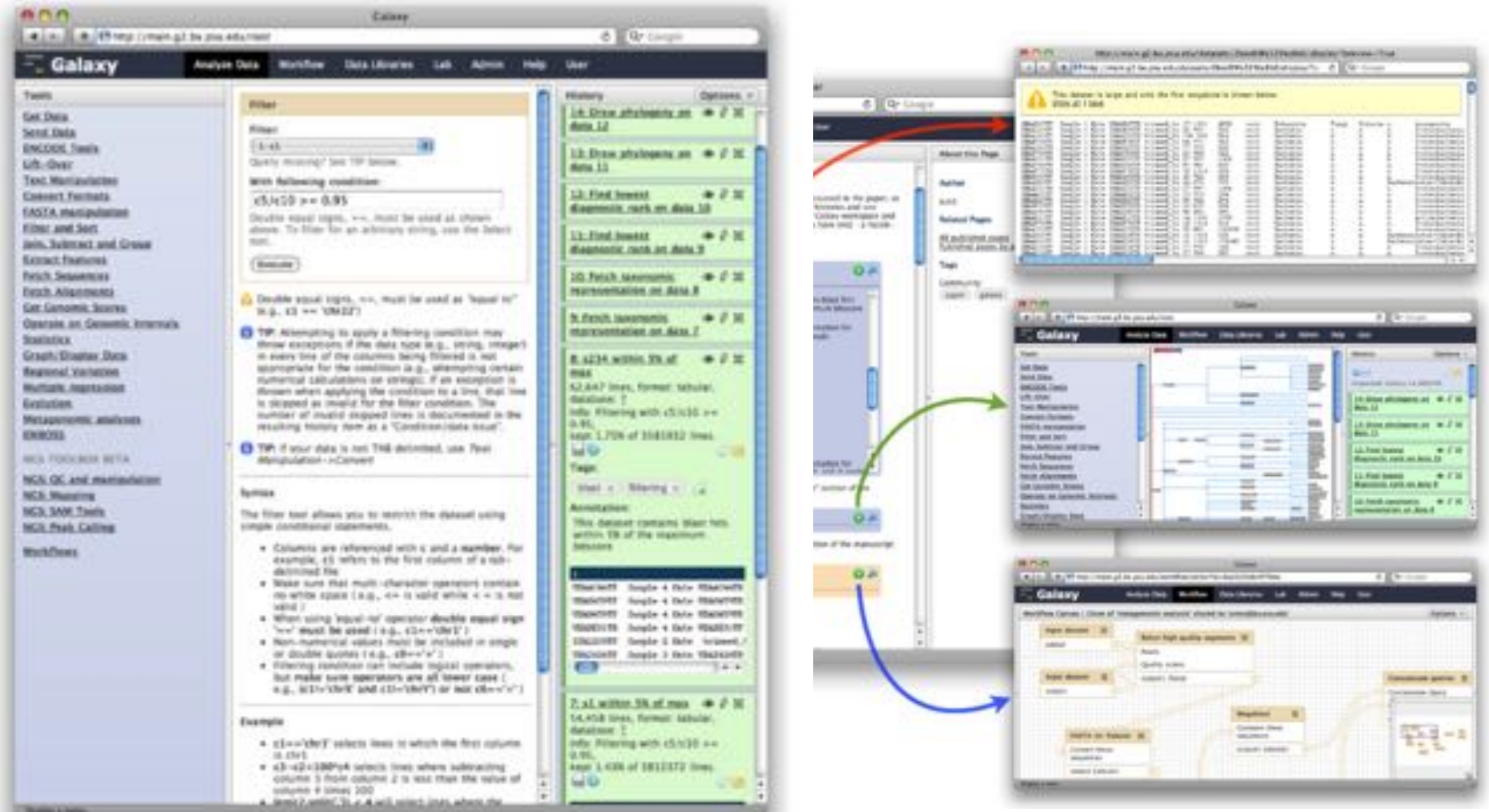
http://meetings.cshl.edu/meetings.html

Journals (RSS feeds and eTOC available)

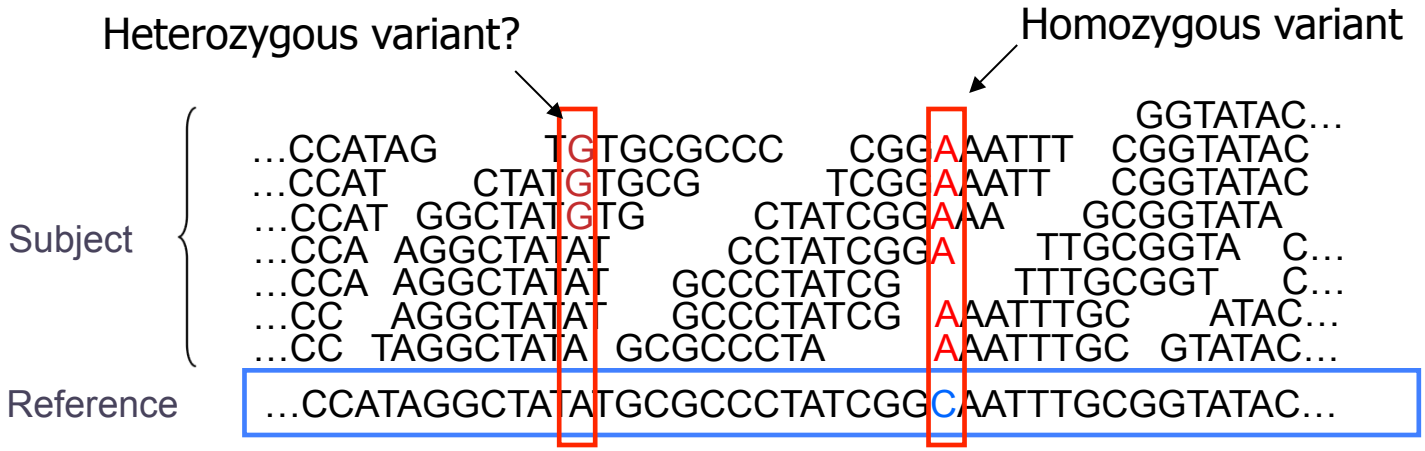
Bioinformatics	Genome Biology	Genome Research
Nature	Nature Biotechnology	Nature Methods
PNAS	PLoS Biology	Science

Galaxy

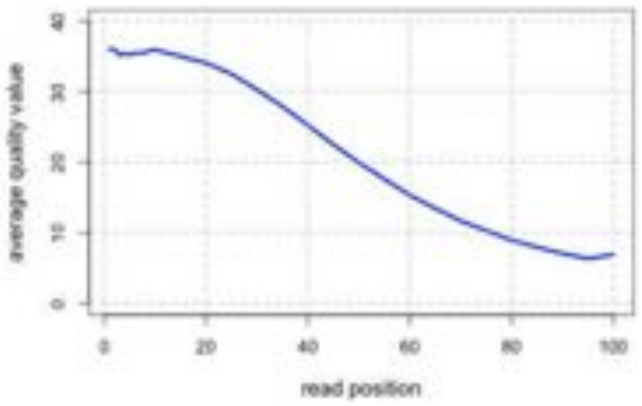
http://usegalaxy.org http://genomics.cshl.edu



Genotyping



- Sequencing instruments make mistakes
  - Quality of read decreases over the read length

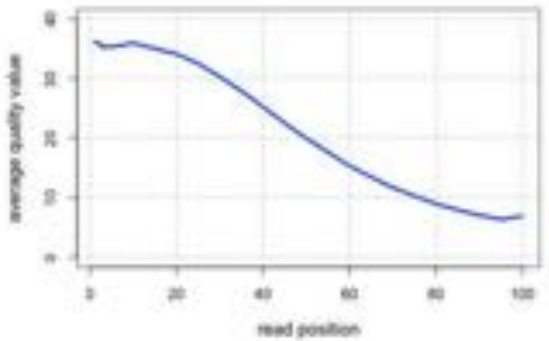


- A single read differing from the reference is probably just an error, but it becomes more likely to be real as we see it multiple times
  - Often framed as a Bayesian problem of more likely to be a real variant or chance occurrence of N errors
  - Accuracy improves with deeper coverage

Illumina Quality

QV	p_{error}
40	1/10000
30	1/1000
20	1/100
10	1/10

$$Q_{\text{sanger}} = -10 \log_{10} p$$

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  SSSSSSSSSSSSSSSSSSSSSSSSSSSSSSSSSS..................................................
  ..........................XXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXXX..............
  ..............................IIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIIII..................
  ...............................JJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJJ..................
  LLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLLL..............................................
  !"#$%&'()*+,-./0123456789:;<=>?@ABCDEFGHIJKLMNOPQRSTUVWXYZ[\]^_`abcdefghijklmnopqrstuvwxyz{|}~
  |                         |    |        |                              |                     |
 33                        59   64       73                            104                   126

 S - Sanger        Phred+33,  raw reads typically (0, 40)
 X - Solexa        Solexa+64, raw reads typically (-5, 40)
 I - Illumina 1.3+ Phred+64,  raw reads typically (0, 40)
 J - Illumina 1.5+ Phred+64,  raw reads typically (3, 40)
     with 0=unused, 1=unused, 2=Read Segment Quality Control Indicator (bold)
     (Note: See discussion above).
 L - Illumina 1.8+ Phred+33,  raw reads typically (0, 41)
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http://en.wikipedia.org/wiki/FASTQ_format

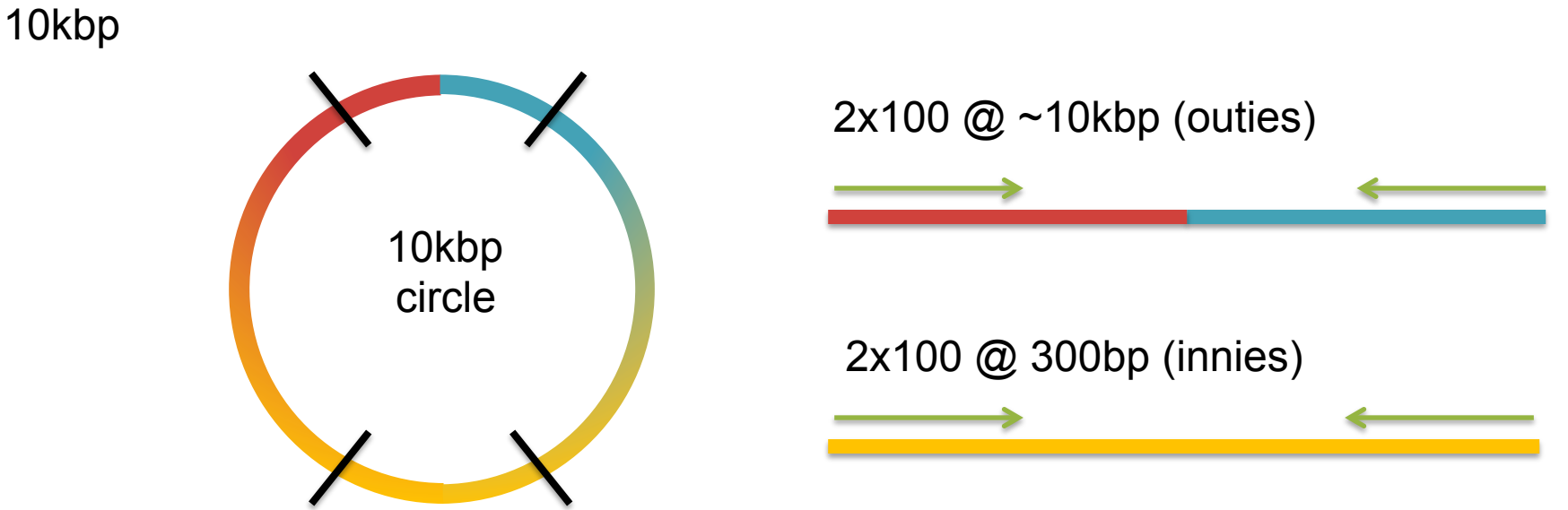
Paired-end and Mate-pairs

Paired-end sequencing

- Read one end of the molecule, flip, and read the other end
- Generate pair of reads separated by up to 500bp with inward orientation

Mate-pair sequencing

- Circularize long molecules (1-10kbp), shear into fragments, & sequence
- Mate failures create short paired-end reads



Galaxy Exercise

1. Download data:
   - http://schatzlab.cshl.edu/teaching/exercises/mapping/mapping.tgz

2. Unpack and upload to Galaxy
   - Set fastq type to fastqillumina of reads

3. Map with Bowtie for Illumina
   - Aligns the reads to the reference genome

4. SAM-to-BAM
   - Converts from ASCII text file to interval representation

5. Coverage Plot of BAM
   - Mapping Statistics

6. Call variants with FreeBayes
   - Print Stats (search vcf)

Other Resources

Resource	URL	Description
Google	http://www.google.com	Internet Search
Google Scholar	http://scholar.google.com/	Literature Searches
SeqAnswers	http://seqanswers.com/	Bioinformatics Forum
Wikipedia	http://www.wikipedia.org/	Overview on anything
Circos	http://circos.ca/	Circular Genome Plots
GraphViz	http://www.graphviz.org/	Graph Visualization
EndNote	http://endnote.com/	Citation Manager
R	http://www.r-project.org/	Stats & Visualizations
Weka	http://www.cs.waikato.ac.nz/ml/weka/	Data Mining
IGV	http://www.broadinstitute.org/igv/	Read Mapping Viz
Schatz Lab	http://schatzlab.cshl.edu/teaching/	Exercises and Lectures

Questions?

http://schatzlab.cshl.edu