MBB 342

SFU SIMON FRASER UNIVERSITY

INTRODUCTORY GENOMICS AND BIOINFORMATICS

#KeyPrereq

#Genomics #Bioinformatics #DryLab #Techniques

Major topics in genomics and bioinformatics, with integrated discussion of associated ethical/legal/social issues. An overview of laboratory and computer-based methods to study genomes, and their applications. Hands-on computer lab session providing an opportunity to use and experiment with bioinformatics software and databases utilized in genomics and bioinformatics research.

TOPICS

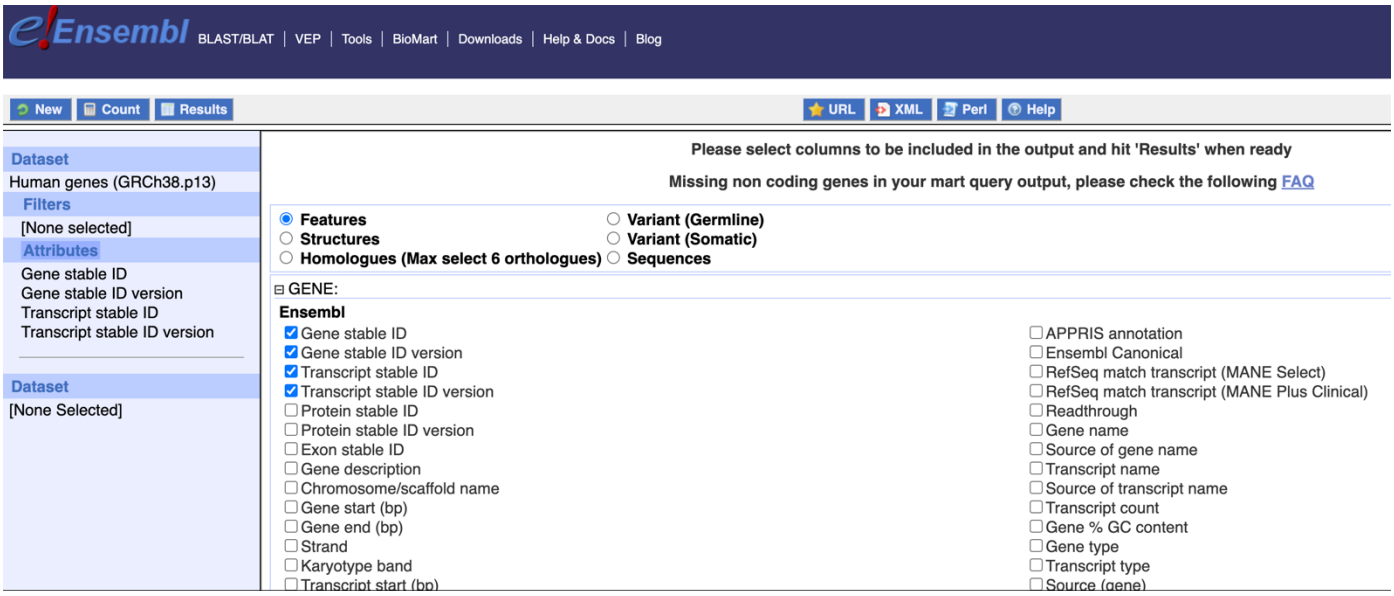
- Obtaining the data: Genome sequencing, genome assembly
- Organizing the data: Gene/genome databases, browsers and searching
- Sequence alignment and sequence similarity search
- Genome assembly and short read mapping
- Human genome variation – SNP, copy number and structural
- Transcriptomics/RNA sequencing, chromatin IP and promoter analysis
- Multiple sequence alignment, intro to evolutionary analysis
- Orthologs, paralogs/gene families, phylogenetic analysis
- Protein, network-based analysis, and Systems Biology
- Microbial genomics and metagenomics of environmental/human microbiomes
- Human genomics and personalized medicine

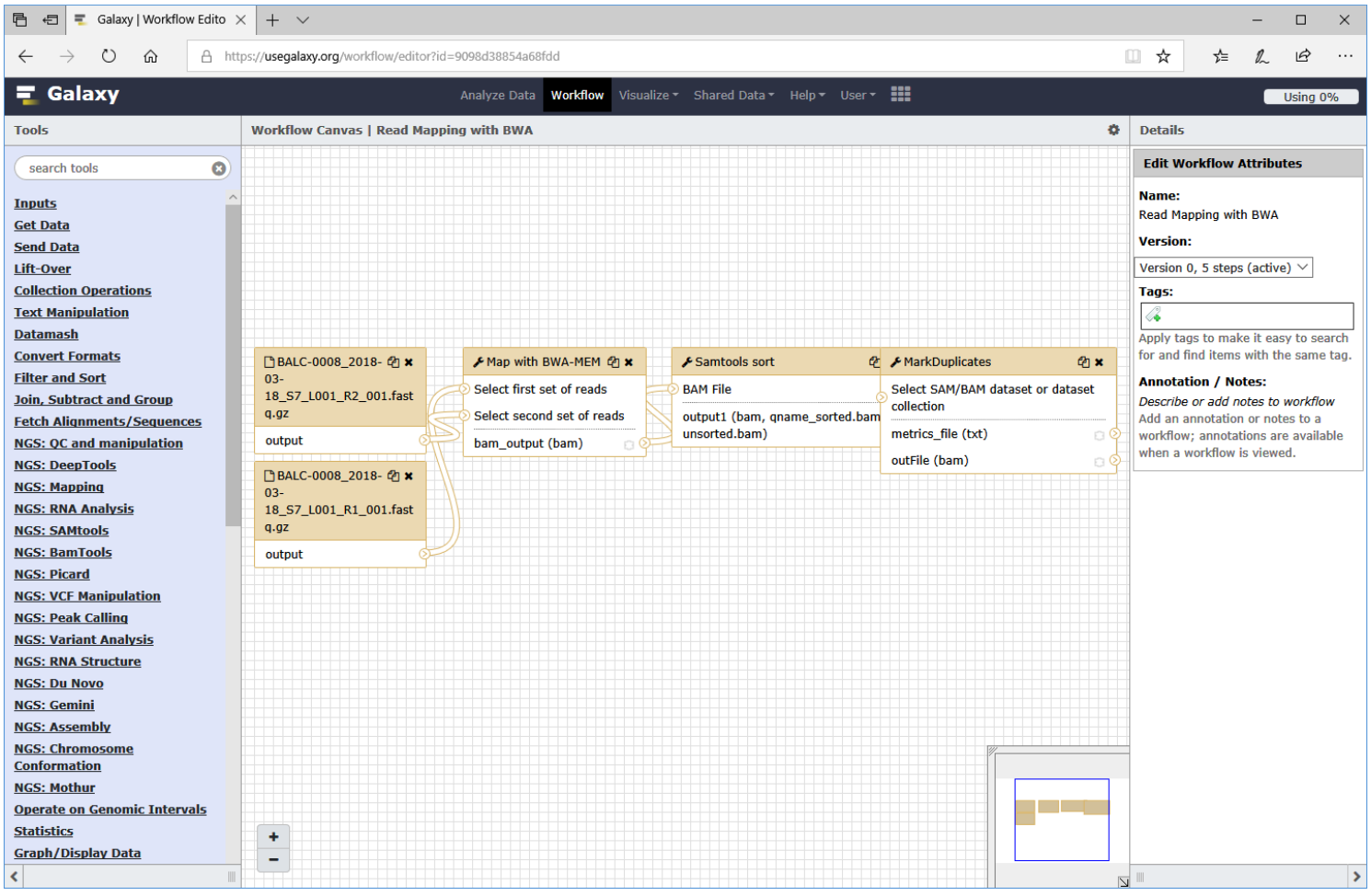
INSTRUCTORS

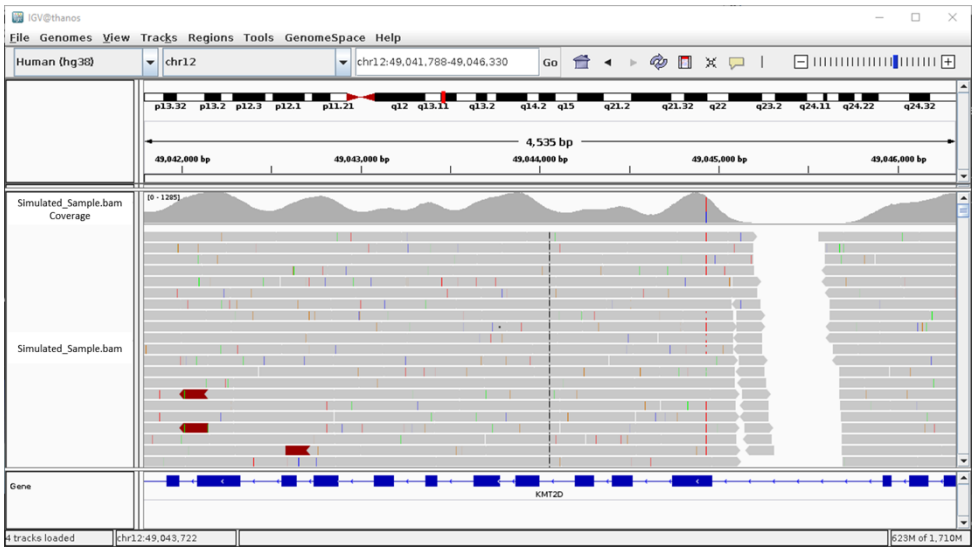
Fiona Brinkman

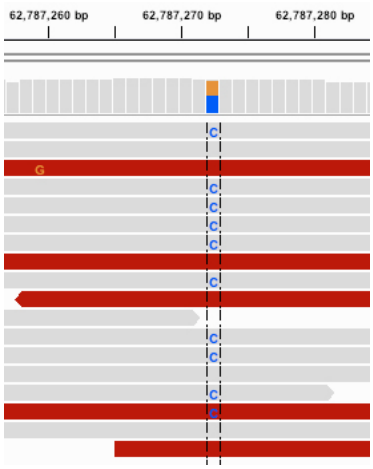
Sophie Sneddon

Bioinformatician, video game enthusiast, cat person

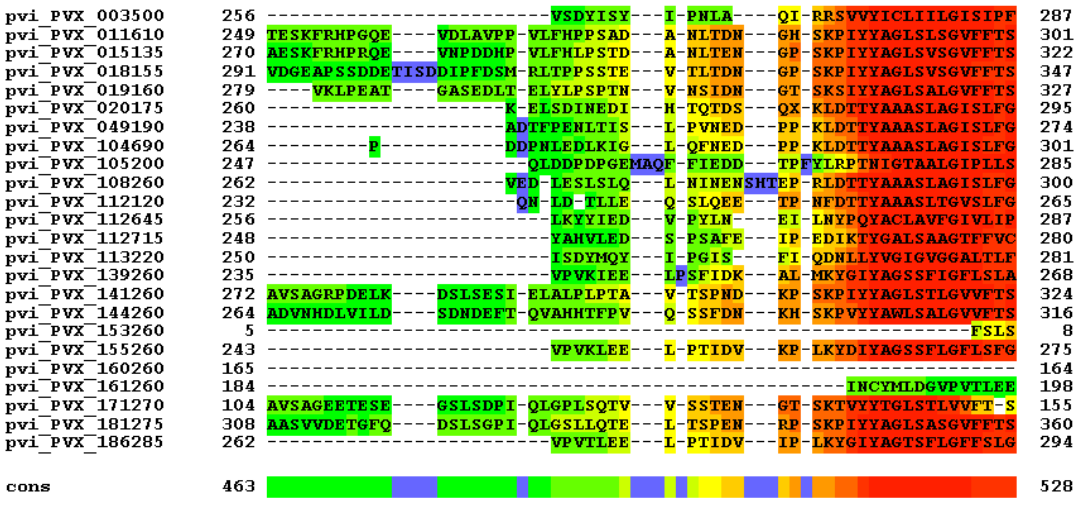

e!Ensembl
BLAST/BLAT | VEP | Tools | BioMart | Downloads | Help & Docs | Blog
New
Count
Results
URL
XML
Perl
Help
Dataset
Human genes (GRCh38.p13)
Filters
[None selected]
Attributes
Gene stable ID
Gene stable ID version
Transcript stable ID
Transcript stable ID version
Dataset
[None Selected]
Please select columns to be included in the output and hit 'Results' when ready
Missing non coding genes in your mart query output, please check the following FAQ
Features
Structures
Homologues (Max select 6 orthologues)
Variant (Germline)
Variant (Somatic)
Sequences
GENE:
Ensembl
Gene stable ID
Gene stable ID version
Transcript stable ID
Transcript stable ID version
Protein stable ID
Protein stable ID version
Exon stable ID
Gene description
Chromosome/scaffold name
Gene start (bp)
Gene end (bp)
Strand
Karyotype band
Transcript start (bp)
APPRIS annotation
Ensembl Canonical
RefSeq match transcript (MANE Select)
RefSeq match transcript (MANE Plus Clinical)
Readthrough
Gene name
Source of gene name
Transcript name
Source of transcript name
Transcript count
Gene % GC content
Gene type
Transcript type
Source (gene)


Galaxy | Workflow Edito
https://usegalaxy.org/workflow/editor?id=9098d38854a68fdd
Galaxy
Analyze Data
Workflow
Visualize
Shared Data
Help
User
Using 0%
Tools
search tools
Inputs
Get Data
Send Data
Lift-Over
Collection Operations
Text Manipulation
Datamash
Convert Formats
Filter and Sort
Join, Subtract and Group
Fetch Alignments/Sequences
NGS: QC and manipulation
NGS: DeepTools
NGS: Mapping
NGS: RNA Analysis
NGS: SAMtools
NGS: BamTools
NGS: Picard
NGS: VCF Manipulation
NGS: Peak Calling
NGS: Variant Analysis
NGS: RNA Structure
NGS: Du Novo
NGS: Gemini
NGS: Assembly
NGS: Chromosome Conformation
NGS: Mothur
Operate on Genomic Intervals
Statistics
Graph/Display Data
Workflow Canvas | Read Mapping with BWA
BALC-0008_2018-03-18_S7_L001_R2_001.fastq.gz
output
BALC-0008_2018-03-18_S7_L001_R1_001.fastq.gz
output
Map with BWA-MEM
Select first set of reads
Select second set of reads
bam_output (bam)
Samtools sort
BAM File
output1 (bam, qname_sorted.bam unsorted.bam)
MarkDuplicates
Select SAM/BAM dataset or dataset collection
metrics_file (txt)
outFile (bam)
Details
Edit Workflow Attributes
Name:
Read Mapping with BWA
Version:
Version 0, 5 steps (active)
Tags:
Apply tags to make it easy to search for and find items with the same tag.
Annotation / Notes:
Describe or add notes to workflow
Add an annotation or notes to a workflow; annotations are available when a workflow is viewed.


IGV@thanos
File Genomes View Tracks Regions Tools GenomeSpace Help
Human (hg38)
chr12
chr12:49,041,788-49,046,330
Go
4,535 bp
Simulated_Sample.bam Coverage
Simulated_Sample.bam
Gene
KMT2D
4 tracks loaded
chr12:49,043,722
623M of 1,710M


62,787,260 bp
62,787,270 bp
62,787,280 bp


Structure Summary
3D View
Annotations
Experiment
Sequence
Genome
Versions
Biological Assembly 1
1HEW
REFINEMENT OF AN ENZYME COMPLEX WITH INHIBITOR BOUND AT PARTIAL OCCUPANCY. HEN EGG-WHITE LYSOZYME AND TRI-N-ACETYLCHITOTRIOSE AT 1.75 ANGSTROMS RESOLUTION
PDB DOI: 10.2210/pdb1HEW/pdb
Classification: HYDROLASE(O-GLYCOSYL)
Organism(s): Gallus gallus
Mutation(s): No
Deposited: 1992-01-20 Released: 1994-01-31
Deposition Author(s): Cheetham, J.C., Artymiuk, P.J., Phillips, D.C.
Experimental Data Snapshot
Method: X-RAY DIFFRACTION
Resolution: 1.75 Å
R-Value Observed: 0.229
wwPDB Validation
Clashscore
Ramachandran outliers
Sidechain outliers
This is version 2.0 of the entry. See complete history.


pvi_PVX_003500 256 287
pvi_PVX_011610 249 301
pvi_PVX_015135 270 322
pvi_PVX_018155 291 347
pvi_PVX_019160 279 327
pvi_PVX_020175 260 295
pvi_PVX_049190 238 274
pvi_PVX_104690 264 301
pvi_PVX_105200 247 285
pvi_PVX_108260 262 300
pvi_PVX_112120 232 265
pvi_PVX_112645 256 287
pvi_PVX_112715 248 280
pvi_PVX_113220 250 281
pvi_PVX_139260 235 268
pvi_PVX_141260 272 324
pvi_PVX_144260 264 316
pvi_PVX_153260 5 8
pvi_PVX_155260 243 275
pvi_PVX_160260 165 164
pvi_PVX_161260 184 198
pvi_PVX_171270 104 155
pvi_PVX_181275 308 360
pvi_PVX_186285 262 294
cons 463 528