Hellenic Association of Medical Geneticists

A UCSC Genome Browser workshop
Athens, 3-4th March 2019

Hands-on training by Dr. Robert Kuhn,
Associate Director of UCSC Genome Browser, Training and Outreach

Sunday 3rd March, 2019

9:00-9:15 Welcome
9:15-9:45 An introduction to NGS technologies
9:45-10:15 NGS An introduction to Bioinformatics

10:15 - 11:30
Introduction to the UCSC Genome Browser - features and navigation.
A guided tour of the basic parts of the UCSC Genome Browser:
* General overview of the Browser, what it is, history
* Display configuration
* Track settings
* Exporting Browser images for publication
* Searches: for regions/positions, gene names, motifs, other keywords
* Searches: BLAT, short match track for sequences
* Searches: PCR for primers
* Demo of example searches: Gene of interest, regions of interest, dataset/publication of interest
* Navigation: drag-and-zoom, drag-reorder tracks, switching genome assemblies and organisms via homology
* Keeping track of your thinking by saving and sharing sessions

11-30 - 11:45 Coffee break

11:45 - 13:00 More Genome Browser - data
A short tour of our data
* Starting points for browsing our data:
  * genomic position
  * sequence
  * gene
  * HGVS nomenclature
* Data types
  * variants -- CNVs and SNPs, benign and pathogenic
  * comparative genomics -- synteny and evolution
  * publications / gene interactions
  * proteomics
* How to find more info about a track or track item

* Table Browser and extracting data
* Custom tracks - uploading and displaying your own data in the Browser
* Custom track examples - BED files. Sequences data: coverage (BAM files), variant calls (VCF), RNA-seq (wiggle)
* Public Assembly Hubs
* How to download our data
  * How to get DNA for the region you are viewing
  * Getting gene sequences using the Table Browser

13:00 - 14:00 Lunch

14:00 - 17:00
Cementing your knowledge: Working through problem sets
One-on-one consultation/troubleshooting
Examples of comparative genomics, changes between assemblies, evolution

Monday 4th March, 2019

9:00 -10:30 Getting more out of the Browser
* Multi-Region mode
  * Exon-only display
  * Viewing regions of your own choice, including discontinuous (e.g., pathways)
  * Alternate haplotypes in human genome assemblies
* Public sessions - sharing your data with everyone
* Table Browser advanced examples:
  * Intersection
  * Filtering
  * Finding the table that goes with your track
* Track Hubs
  * How to make your own track hub
* Assembly Hubs
  * Make a track hub for your genome of interest
* New feature: Track collections

10-30 - 10:45 Coffee break

10:45 - 12:30 Browsing variant data and sensitive data
* Understanding various SNP tracks
* The Variant Annotation Integrator: predicting biochemical consequences of short variants
* Genome Browser in a Box: Hosting a local copy of the Genome Browser on your laptop
  * GBiB as Browser inside your firewall
  * Using GBiB and Assembly hubs to explore sensitive data

12:30 - 13:30 Lunch

13:30 - 16:00
Cementing your knowledge: Advanced problem session to work on example problems
One-on-one consultation/troubleshooting
Using the Browser in teaching genetics and molecular biology

16:00-16:30 Closing discussion