เรื่อง "Next-Gen Sequencing 101: Clinical Exome & Target Region Sequencing Workshop"

ระหว่างวันจันทร์ – พุธ ที่ 17–19 กุมภาพันธ์ 2557

ณ ห้องคอมพิวเตอร์ 317 ชั้น 3

อาคารเรียนใหม่ โรงเรียนพยาบาล โรงพยาบาลรามาธิบดี

เนื้อหาหลักสูตร

Day 1: February 17, 2014	
8:00-8:45	Registration
8:45-9:00	Opening ceremony: *Dean of Faculty of Medicine, Ramathibodi Hospital, Mahidol University*
9:00-12:00	(Lecture1) The completion from small pathogen genomes to whole human genome sequencing in less than 3 hours -Using NGS for Exome sequencing -Using NGS for Transcriptome sequencing -Using NGS for Whole Human Genome sequencing -Using NGS for Target region sequencing. Inherited Disease and Cancer Panel of target genes. -Using NGS for Microbial sequencing ***Instructors: Wasun Chantratita, Ekawat Pasomsub, Jeerawat Nakkuntod, Chuphong Thongnak***
12:00 - 13:00	**Lunch Break**
13:00-15:00	Lab Demonstration **1** Demonstration 1 NGS wet lab **(Gr.1 13:00-15:00, Gr.2 15:00-17:00)** - Whole human genome sequencing -Rapid exome, transcriptome, targeted amplicon sequencing using semiconductor sequencing technology - Primer design tool to create custom, ultrahigh-multiplex primer pools for next generation sequencing. -Next generation sequencing comprehensive inherited disease gene panels. - Next generation sequencing comprehensive cancer gene panels. - HIV-1, HBV, and HCV deep sequencing and interpretation ***Instructors: Chutatip Srichunrusami, Pareena Janchompoo, Wichuda Narkpoung, Yanika Keeratiwongsa, Suthee Benjaphokee, Haiyan Guo, Kristian Ridley***

15:00-17:00 Lab Demonstration **2**

Demonstration 2 Point-and-Click software programs for NGS data analysis tools **(Gr.1 15:00-17:00, Gr.2 13:00-15:00)**

- Partek Genomics Suite: Server based RNA-Seq, ChIP-Seq, and Methyl-Seq and DNA-Seq.

- DNASTAR VS Golden Helix VS Enlis Genomics: Server based NGS genome analysis software suites.

- Ion reporter: Cloud based-automated variant analysis and driver mutation identification for clinical research.

Instructors: Ekawat Pasomsub, Chuphong Thongnak, Nipaporn Sankuntaw , Life technologies, and Partek specialists)

Day 2: February 18, 2014

9:00-12:00 **(Lecture 2)** Finding genes for Mendelian disorders. Past and present

- Mendelian disorders. Definition. Short repetition of the main terms

- Overview of the past methods and approaches. Examples of it successes and limitations. Need for new methods to resolve the remaining cases

- NGS technology as one of the main breakthrough in medical genetics of the past decade

-Exome capture technology. New method in finding genes for Mendelian disorders.

Instructor: Marianna Bevova

(Lecture 3) Exome sequence. Overview of the workflow: from samples to data analysis

- How to choose which samples to sequence?

- DNA isolation and exome capture

- NGS technique overview (Illumina technology as an example)

- Data analysis

- **QC of the data**
- **Alignment of the reads**
- **Variant calling**
- **Variant filtering**
- **Data interpretation**

Instructor: Marianna Bevova

(Lecture 4) Initial QC of the data. Alignment. Variant calling

	- Importance of QC in next generation sequencing data **- Main parameters to take into account** **- FastQC program for data quality check** **- Mapping sequence reads** **- Quality control of aligned data** **- Mapping files formats** **- Tools for calling SNPs and structural variants** **- Visualizing of the data. IGV software and USCS genome browser** ***Instructor: Victor Guryev***
12:00 - 13:00	**Lunch Break**
13:00 - 16:30	**Practical session 1** **1. FastQC software - quality of the sequence data** **2. Mapping the sequence reads** **3. Variant calling** **4. Data visualization (IGV, UCSC genome browser)** ***Instructors: Victor Guryev, Marianna Bevova***

Day 3: February 19, 2014	
9:00-11:00	(Lecture 5) Structural variant calling in whole exome sequence *Instructor: Victor Guryev*

(Lecture **6**) Annotation, filtering and prioritization of variants

- Databases to annotate the variants
  - Identification of variants (nomenclature, standard databases)
  - Frequencies (population frequency, control dataset)
- Algorithms and softwares for functional predictions. Overview. Limitations
  - Functional modification of the proteins
  - Species conservation
- Other annotations
  - Literature, knowledge
  - Transcription/posttranscriptional effects
- Filtering strategies to find the causal variant. Examples of the different scenarios.

Instructor: Marianna Bevova

11:00 - 12: 00	**Practical session 2** **Finding the causal variant in the whole exome sequence data** ***Instructors: Marianna Bevova, Victor Guryev***
12:00 - 13:00	**Lunch Break**
13:00-14:00	**Practical session (continuation)**
14:15 - 16.30	**(Lecture 7) Variant found- what next?** **Main steps to prove causality of the variant. Integration exome data with other genomic data** *Instructor: Marianna Bevova*
	(Lecture 8) Exome & Target region sequencing of inherited diseases and familial cancers. *Instructor: Victor Guryev*
	(Lecture 9) Ethical issues. Incidental and secondary findings in exome data. Going back to the family *Instructor: Marianna Bevova*
	(Lecture 11) Exome & Target region sequencing as a strategy to find gene for Mendelian disorders and cancer Advantages and limitations. From research to diagnostics. Conclusions. *Instructor: Marianna Bevova*