AGILENT NGS SEMINAR

THOMAS JEFFERSON UNIVERSITY
HAMILTON 505
TUESDAY, APRIL 5, 2016
1:30PM - 4:00PM

1:30 - 2:15pm **NGS 101 and Target Enrichment NGS Applications in Cancer Research**

Josh Wang, Ph.D, Field Application Scientist, Agilent Technologies, Inc.

Next-generation sequencing (NGS) has become an indispensable genetic tool in basic and clinical research applications. In this presentation we will cover the basic NGS concepts including NGS sequencers, NGS library preparations, NGS sequence reads and data analysis as well as NGS target enrichment focusing on a subset of regions of interest instead of whole genome or whole transcriptome. We will use cancer research applications to illustrate the power of NGS target enrichment methodology on cancer gene mutation detection at high sensitivity (down to < 1% allele frequency) and cancer predisposition gene screening.

2:15 - 3:00pm **Agilent SureDesign NGS Tools: Navigation and Understanding Advanced Features**

Josh Wang, Ph.D, Field Application Scientist, Agilent Technologies, Inc.

SureDesign is a free web-based DNA application design tool from Agilent, the leader in NGS target enrichment and array CGH. Its suite of applications include SureSelect, HaloPlex HS, CGH array, custom FISH and CRISPR guide DNA etc. With the NGS applications, SureSelect and HaloPlex HS, one can design a custom library with a list of genes, exons, SNPs, and/or contiguous regions at ease to detect variants, indels, copy number variations and translocations. In this presentation we will focus on navigating SureDesign user interface, specifically on SureSelect and HaloPlex HS, with live demos. Furthermore, we will also discuss advanced design parameters such as tiered masking, probe boosting option, maximize coverage and FFPE optimization options etc to help users understand various strategies to enhance a custom design performance.

3:00 - 4:00pm **Integrated Genomic Analysis of Cancer Genomes for the Management of Pediatric and Adult Cancers**

Marilyn M. Li, M.D, Division of Genomic Diagnostics, The Children's Hospital of Philadelphia, Professor of Pathology & Laboratory Medicine, University of Pennsylvania, Professor of Internal Medicine, Perelman School of Medicine

Next-generation sequencing (NGS) technologies have revolutionized genomic research by decreasing the cost of sequencing while increasing the throughput. The focus now is on the clinical research applications of NGS technology. Clinical research application of NGS in cancer can detect genetic/genomic alterations that are critical for cancer care. In certain cancers, risk and prognosis can be predicted based on the mutation profile identified by NGS. Many targeted therapies have been developed for cancer patients who bear specific mutations. However, choosing right NGS techniques for appropriate clinical research applications can be challenging, especially in clinical research oncology, where the material for testing is often limited and the turnaround time of testing is frequently constrained to just a few days. Currently, targeted NGS approaches have emerged as the best fit for oncology clinical research. We have developed and validated multiple NGS panels that allow the detection of point mutations, small indels, copy number variations, and novel fusion genes in different cancers, as well as mutations associated with cancer predispositions. These panels have been applied to thousands of cases and have provided critical genomic information for precision cancer care. Currently, whole exome and whole genome sequencing are mostly used in cancer research. As the cost of running NGS-based test continues to decrease and better software for NGS data analysis continues to improve, clinical research application of whole exome, whole genome, and whole transcriptome sequencing in cancer care is just a matter of time.

Agilent Technologies