Roche Seminar Information

The 13th International Congress of Human Genetics

Luncheon Seminar 18（LS18）

Theme

Transforming the state of genomic biomarker discovery and diagnostic testing with high-throughput long-read DNA sequencing technologies

Speaker

Chairman, Department of Genetics and Genomic Sciences
Founding Director, Icahn Institute for Genomics and Multiscale Biology
Icahn School of Medicine at Mount Sinai, New York
Eric Schadt, PhD

Moderator

Scientific Director
Roche Sequencing Solutions
William LaRochelle PhD

Date

April 7（Thu）, 2016

11:30～12:30

Venue

Kyoto International Conference Center 2F # Room A

422 Iwakuraosagi cho Sakyo-ku, Kyoto-shi Kyoto, 606-0001, Japan

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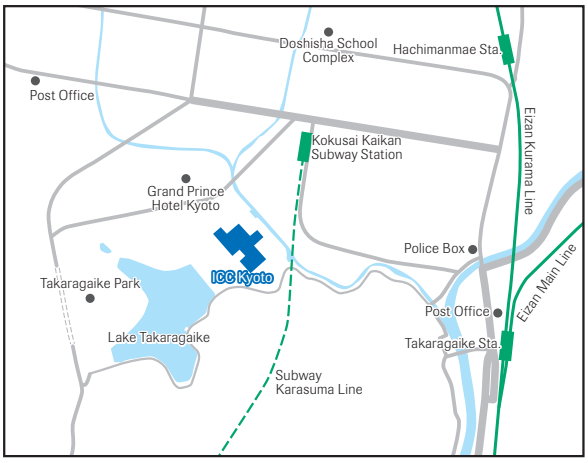
The 13th International Congress of Human Genetics
Sponsored by Roche Diagnostics K.K.



Traffic Access
[Train] Subway　5 minutes from Kokusai Kaikan Subway Station by walk.

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Transforming the state of genomic biomarker discovery and diagnostic testing with high-throughput long-read DNA sequencing technologies

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Founding Director, Icahn Institute for Genomics and Multiscale Biology
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Eric Schadt, PhD

Abstract

One of the primary goals of precision medicine is the aggregation and interpretation of deep, longitudinal patient-specific data in the context of the digital universe of information, using advanced predictive analytics to better diagnose and treat patients, even down to tailoring individualized treatments to patients as happens today in areas such as novel vaccine development targeted to individual tumors. Central to the implementation of precision medicine strategies is the accurate and comprehensive characterization of DNA variation in the human genome that helps define disease risk, severity, progression, and drug response. However, to date, given the widespread use of short-read DNA sequencing technologies for characterizing DNA variation, variation central to disease processes, wellness, and drug response are systematically missed given the limitations in short-read sequencing technologies to assay complex repeat structures throughout the genome associated with disease and protection against disease.

To address these limitations and provide a more comprehensive assessment of genome variation associated with disease and drug response, we have employed the SMRT sequencing technology from Pacific Biosciences to access what has previously been considered unresolvable and even unsequencable genomic regions through unbiased, very long read sequence reads that typically span many thousands or even tens of thousands of bases. In fact, recent advances in the PacBio SMRT technology now available in their Sequel platform have dramatically increased the sequencing throughput, making it possible for the first time to tackle large-scale population-based studies using long-read sequencing. Beyond the more general, genome-wide characterizations of variation that are enabled with this new technology, we have developed high-grade clinical assays for routine genetic testing in complex gene regions associated with disease, including long-range, high fidelity sequence capture in tandem with long-read sequencing in BRCA1/2 genes for assessing heritable cancer risk, CYP2D6 for more comprehensive assessment of response to a wide range of drugs including tamoxifen, and the HLA region to more comprehensively characterize variation in HLA genes in support of our novel vaccine development strategies for immunotherapy in different cancers currently in clinical trials. Beyond these human genome characterizations, we have also employed these long-read strategies to unambiguously resolve complex, heterogeneous mixtures of viruses such as within-host virus diversity in patients infected with influenza A virus, enabling for the first time the ability to characterize viral variants that achieve sustainable transmission patterns in new hosts.

Given these capabilities to more comprehensively and accurately assess genetic variation in pathologically relevant genome regions in support of clinical decision making, the potential exists to characterize the health of an individual at a far deeper level than has been possible before. Better characterization of an individual's genome in the context of other molecular, physiologic, and environmental data collected on them will lead to more precise diagnoses, treatments, and preventative strategies for disease at a highly personalized level.

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