

Next-Generation Sequencing Data Management

April 20, 2010

8:00 am – 4:00 pm World Trade Center, Boston, MA

This workshop will present real-world customer experiences straight from the trenches. You'll get practical information about the analysis, assessment, design, implementation, testing, and support needed to bring a research organization from technology adoption to publication in the next-gen world.

> Held in conjunction with CHI's Ninth Annual



Cambridge Healthtech Institute 250 First Avenue, Suite 300 Needham, MA 02494 Telephone: 781-972-5400 or toll-free in the U.S. 888-999-6288 Fax: 781-872-5425 | www.healthtech.com

Enabling Technology. Leveraging Data. Transforming Medicine. April 20-22, 2010 | World Trade Center, Boston, MA Bio-ITWorldExpo.com

Workshop Agenda

7:00-8:00 am Pre-Conference Workshop Registration and Morning Coffee

8:00 Workshop Chairperson's Remarks

William Van Etten, Ph.D., Founding Partner, BioTeam, Inc.

8:15 Third Generation Sequencing and Interpretation Technologies for Open-Access Whole Genome-Phenome Data

George Church, Ph.D., Professor, Genetics, Harvard Medical School; Director, Center for Computational Genetics

The cost of sequencing has plummeted a million-fold in 6 years. Suddenly individuals possess their own genomic and electronic health records, raising a plethora of issues in safeguarding genomic and clinical data. We have launched the Personal Genome Project, which explores a different approach – gaining truly informed consent in advance (after the subject passes an exam) with the understanding of full disclosure. We have developed a community resource of software and data for interpreting genome sequence data called Trait-omatic, as well as open-source community tools for webbased computer adaptive phenotyping.

8:45 Don't Move the Data—Move the Jobs! Phil Butcher, Head of IT, The Wellcome Trust Sanger Institute

As we move steadily forward with sequencing technologies, we continue to face increasing challenges in data management. New projects are requesting Petabytes of storage capacity to add to the 4 Petabytes already installed at the Sanger Institute. Major sequencing centers like us need to think about the underlying infrastructure and the way we manage the storage to keep production moving. This talk will present our current thinking and strategies for managing these large data capacities.

9:15 Epigenome-Wide Association Studies

John M. Greally, M.B.A., Ph.D., Director, Center for Epigenomics, Chief, Division of Computational Genetics, Department of Genetics, Albert Einstein College of Medicine

To allow investigators to study the role of epigenomic dysregulation in human disease, a number of hurdles have to be overcome. These include study design, application of suitable molecular assays, and data management and analysis. While many of these issues reflect those for genome-wide association studies (GWAS), epigenomic studies have unique challenges that require specific solutions. We describe how we have created an infrastructure designed to facilitate testing the role of epigenomic dysregulation in human diseases.

9:45 Refreshment Break

10:00 Enabling Plant Research with Next-Generation Sequencing

Rod Wing, Director, Arizona Genomics Institute, University of Arizona

10:30 Enabling Next-Generation Sequencing

Giles Day, Senior Director, R&D Informatics, Pfizer, Inc.

The growth of sequencing data in the modern enterprise is now a well documented issue. This presentation will focus on the strategies and software that have been used within Pfizer's Biotherapeutics Division to successfully support sequencing strategies for both internally and externally generated data. Experimental design, data delivery, integration and analysis will be discussed and how these obstacles have been overcome.

11:00 Scaling storage Capacity in Step with Scanner Throughput; Solutions for Maximizing Research and Minimizing Storage Expenditure



Carter George, Vice President, Products, Ocarina Networks

In all Bioinformatics disciplines, rich data is the platform from which all discovery is made, and rapid throughput growth of highly anticipated next-generation scanning systems means faster time-to-discovery. However, with each increase in scanner throughput comes a corresponding requirement for additional storage. Because scanner throughput is out-pacing disk system density, bioinformatics organizations may be allocating more and more of their budgets to IT and less to research. The deployment of cheaper disk is attractive, but imposes higher administrative costs. This presentation will review various solutions to keeping IT expenditures in check, without sacrificing on enterpriseclass reliability and scalability features. In particular Carter will examine the latest in deduplication and compression technologies, their tradeoffs and intended use models, and how they integrate with existing storage systems.

11:30 Finding Clarity in the Cloud— Separating Fact from Fiction in Cloud Computing for Life Sciences



Paul Rutherford, Chief Technology Officer, Isilon Systems

IDC recently stated it expects revenue from Cloud IT services to increase by a 26% CAGR through 2013, compared to only 4% for traditional IT services, demonstrating there is actual momentum driving the Cloud's hype. However, in the same study, respondents noted security, availability, performance and concern the "pay what you use model" may actually be more expensive than building your own infrastructure, as serious impediments to cloud adoption. It's no wonder then that many of us are still unsure what to make of the Cloud – fact or fiction?

As with many new IT trends, the answer lies somewhere in between. However, the outlook need not remain so cloudy. Isilon CTO Paul Rutherford will use this presentation to detail exactly what the Cloud is – and isn't – and what best practices life science organizations can follow to ensure they make the right IT investment.

As part of this presentation, Rutherford will address the following questions: What is the Cloud? Private vs. Public?

Will the Cloud save money? Will the Cloud accelerate application performance? Will the Cloud increase IT efficiency? The answers may surprise you.

1:30 Visualization of Next-Generation Data

Michael Reich, Director of Cancer Informatics Development, Broad Institute of MIT and Harvard

Visualization plays an important role in assimilating the large, muti-modal datasets produced by genome characterization efforts and other high-throughput research initiatives. To be maximally effective, tools must flexibly integrate genomic, functional, and phenotypic data from a variety of platforms, and efficiently handle large datasets. To address these critical challenges, the Broad Institute of MIT and Harvard developed the Integrative Genomics Viewer (IGV), a flexible, scalable, high-performance tool for the concurrent visualization of multiple large scale datasets. The IGV supports integrated views of any data type that can be mapped to genomic coordinates, including, but not limited to, next-generation sequence data, copy number, gene expression, RNAi, ChIP-seq, RNA-seq, and genome annotations. A particular strength of IGV is its ability to handle very large datasets, for example deep coverage sequence data from whole genome scans, or high-resolution copy number data from thousands of samples, on standard desktop computers. IGV permits the user to move seamlessly across these datasets at any level of detail, and quickly zoom out to whole genome or drill down to base pair resolution. The ability to dynamically and flexibly integrate multiple data modalities and view them at any scale allows the user to elucidate complex biological relationships that would not otherwise be apparent.

2:00 How to Overcome the 100 Miles between Petabases and Petabytes

Jurgen Eils, Bioinformatic Database Group Leader, German Cancer Research Center

In the new field of NGS (next-generation sequencing) we are currently preparing our IT infrastructure to be ready for next-generation computing. Recently, the Heidelberg University received a grant to build up the largest data storage facility in Germany. The 5-10 petabyte large scale data facility (LSDF) is presently being set up in private partnership with a leading company for storage solutions. From the management and logical perspective, the massive throughput of next generation sequencing requires new concepts and strategies. We will present our strategies and concepts with emphasis on reusability and sustainability for storing and retrieving the comprehensive and exhaustive collection of sequence data together with secondary clinical and histopathological annotation data - and all in accordance with the international cancer genome consortium (ICGC) guidelines.

3:00 Panel Discussion: Industry Overview



3:45 Workshop Chairperson's Closing Remarks *William Van Etten, Ph.D., Founding Partner, BioTeam, Inc.*

Next-Generation Sequencing Data Management

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