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Dear Colleagues and Friends,

In partnership with the Track Chairs and the rest of the Scientific Program Committee, I am delighted to welcome you to the Fifth AMIA Summit on Translational Bioinformatics! Thanks to the number of high-quality submissions, we have assembled a terrific program with tutorials, lectures, panels, and posters that showcase the latest advances in applying informatics to biomedical research and clinical care. This year, we have four tracks covering research that takes us from base pairs to the bedside, with an emphasis on clinical implications of mining massive data-sets, and on bridging the latest multimodal measurement technologies with large amounts of healthcare data.

Your participation in the Summit is essential for the trans-disciplinary innovations required for the success of translational bioinformatics. We hope you find the Joint Summits to be a fruitful experience from which to gain new insights, new collaborators, and perhaps even new hires. At the TBI Summit, you will find a unique blend of expertise from academia, industry, government, and non-profit sectors that will drive the future of translational bioinformatics, and its impact on medicine. With your participation, we continue to advance this research field by creating a unique learning environment that bridges innovations in biomedical research to patient care.

Future Summits and progress in translational bioinformatics depend on your individual and collective engagement. I am honored to have your company and extend a warm welcome to you.

Enjoy the Summit!

Nigam Shah
Chair, TBI Summit Scientific Program Committee
Stanford University

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# TBI SUMMIT AT A GLANCE

## MONDAY, MARCH 19

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tbody>
<tr>
<td>7:30 – 8:30 am</td>
<td>Coffee and Pastries</td>
</tr>
<tr>
<td>7:30 am – 5:00 pm</td>
<td>Registration Open</td>
</tr>
<tr>
<td>8:30 am – 12:00 pm</td>
<td>Tutorials (additional registration required)</td>
</tr>
<tr>
<td></td>
<td>T01: Introduction to Translational Bioinformatics</td>
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<td></td>
<td>T02: Ontology Services for Translational Research in the i2b2 Workbench</td>
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<td>T03: Reusing EHRs for Clinical, Genomic, and Pharmacogenomic Discovery at Vanderbilt and within the eMerge Network</td>
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<td>T04: Introduction to R for Bioinformatics and Biomedicine</td>
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<tr>
<td>10:00 – 10:30 am</td>
<td>Coffee Break</td>
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<tr>
<td>1:30 – 3:00 pm</td>
<td>Opening Session and Keynote Address: Dr. Trey Ideker</td>
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<tr>
<td>3:00 – 3:30 pm</td>
<td>Coffee Break</td>
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<tr>
<td>3:30 – 5:00 pm</td>
<td>Scientific Sessions</td>
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<td>TBI-01: Predictions from Large Datasets</td>
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<td>TBI-02: Disease Informatics</td>
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<td>TBI-03: Mining the EHR</td>
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<td>TBI-04: SaaS-Based Translational Research: The Path to Reality for “Research in the Cloud”</td>
</tr>
<tr>
<td>5:00 – 6:00 pm</td>
<td>Reception</td>
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## NEW THIS YEAR! BULLET PRESENTATIONS!

Poster authors will present a five minute “bullet presentation” of their poster to convey their innovative ideas! Don’t miss this rare opportunity to learn about cutting edge ideas in the field. Bullet presentations will take place after paper and podium abstract presentations sessions.
**TUESDAY, MARCH 20**

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
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<tbody>
<tr>
<td>8:00 – 9:00 am</td>
<td>Coffee and Pastries</td>
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<tr>
<td>8:00 am – 5:00 pm</td>
<td>Registration Open</td>
</tr>
<tr>
<td>9:00 – 10:30 am</td>
<td><strong>Scientific Sessions</strong></td>
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<td>TBI-05: Research Platforms</td>
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<td>TBI-06: Semantic Querying</td>
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<td>TBI-07: Methods for Large Datasets</td>
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<td>TBI-08: Practical Experience with Linked Open Biomedical Data: State of the Art and Future Directions</td>
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<tr>
<td>10:30 – 11:00 am</td>
<td>Coffee Break</td>
</tr>
<tr>
<td>11:00 am – 12:00 pm</td>
<td><strong>Keynote Presentation: Dr. Howard Jacob – “Genome Sequencing – is it ready for the Clinic?”</strong></td>
</tr>
<tr>
<td>12:15 – 1:15 pm</td>
<td>Lunch &amp; Learn - Apixio (Clinical Knowledge Exchange)</td>
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<tr>
<td>1:30 – 3:00 pm</td>
<td><strong>Scientific Sessions</strong></td>
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<tr>
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<td>TBI-09: Clinical Genomics</td>
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<td>TBI-10: Data Mining</td>
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<td>TBI-11: Tools and Communities</td>
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<td>TBI-12: Knowledge Synthesis for in silico Science: Lessons Learned and Future Directions</td>
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<tr>
<td>3:00 – 3:30 pm</td>
<td>Coffee Break</td>
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<tr>
<td>3:30 – 5:00 pm</td>
<td><strong>Scientific Sessions</strong></td>
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<td>TBI-13: Data Integration Methods</td>
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<td>TBI-14: Analyses with Phenotypes</td>
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<td>TBI-15: Detection and Prediction of Adverse Drug Events in Clinical and Molecular Data</td>
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<td>TBI-16: tranSMART: an Open Source Analytical and Data Sharing Informatics Platform Enabling Translational Research</td>
</tr>
<tr>
<td>5:00 – 6:00 pm</td>
<td><strong>Poster Session and Reception</strong></td>
</tr>
</tbody>
</table>
# TBI SUMMIT AT A GLANCE

## WEDNESDAY, MARCH 21

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tr>
<td>7:30 – 8:30 am</td>
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<tr>
<td>7:30 am – 5:00 pm</td>
<td>Registration Open</td>
</tr>
<tr>
<td>8:30 – 10:00 am</td>
<td><strong>Keynote Presentation: Dr. Robert M. Califf</strong></td>
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<tr>
<td>10:00 – 10:30 am</td>
<td>Coffee Break</td>
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<tr>
<td>10:30 am – 12:00 pm</td>
<td><strong>Scientific Sessions</strong></td>
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<tr>
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<td>TBI-17: Integrative Genomics</td>
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<td>TBI-18: Crossing the Omic Chasm: Integrating Omic Data into the EHR</td>
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<td></td>
<td>TBI-19: Late Breaking Session - National Center for Advancing Translational Sciences</td>
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<tr>
<td></td>
<td>CRI-01: Identifying Patients for Clinical Studies from Electronic Health Records: The TREC Medical Records Track</td>
</tr>
<tr>
<td>12:15 – 1:15 pm</td>
<td>Lunch &amp; Learn - Velos (New Clinical Research Tools)</td>
</tr>
<tr>
<td>1:30 – 3:00 pm</td>
<td><strong>Scientific Sessions</strong></td>
</tr>
<tr>
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<td>TBI-20: Big Data</td>
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<tr>
<td></td>
<td>CRI-02: Natural Language Processing Methods for CRI</td>
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<tr>
<td></td>
<td>TBI-21/CRI-03: Informatics Challenges and Solutions for Capturing, Taming, and Analyzing the Data Tsunami: Report of the ACMI 2012 Winter Symposium</td>
</tr>
<tr>
<td></td>
<td>TBI-22: Intellectual Property and Entrepreneurship in Translational Bioinformatics</td>
</tr>
<tr>
<td>3:30 – 5:00 pm</td>
<td><strong>Closing Session and TBI Year in Review: Dr. Russ Altman</strong></td>
</tr>
<tr>
<td>5:00 – 6:00 pm</td>
<td>Joint Summits Reception, Sponsored by Booz Allen Hamilton</td>
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The American Medical Informatics Association designates this educational activity for a maximum of 26.5 AMA PRA Category 1 Credit(s)™. Physicians should only claim credit commensurate with the extent of their participation in the activity.

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As a sponsor accredited by the ACCME, the American Medical Informatics Association requires that everyone who is in a position to control the content of an educational activity disclose all relevant financial relationships with any commercial interest prior to the educational activity. The ACCME considers relationships of the person involved in the CME activity to include financial relationships of a spouse or partner.

Faculty and planners who refuse to disclose relevant financial relationships will be disqualified from participating in the CME activity. For an individual with no relevant financial relationship(s), the participants must be informed that no conflicts of interest or financial relationship(s) exist.
TRACK DESCRIPTIONS

**TRACK 1: CONCEPTS, TOOLS AND TECHNIQUES FOR TRANSLATIONAL BIOINFORMATICS**

Translational informatics research requires the co-ordinated analysis of molecular as well as clinical phenotypic information to understand the pathophysiology or disease as well as to predict responses to therapeutic interventions. This track will focus on methods, tools and techniques ranging from text-mining, cloud computing and Semantic Web Technologies as well as information integration and data warehousing. Presentations will demonstrate case studies, prototype implementations and mature, production grade tools and platforms.

**TRACK 2: INTEGRATIVE ANALYSIS OF MULTI-MODAL MEASUREMENTS**

With over 30 different high-throughput measurement modalities for measuring the disease state at multiple levels, there is constant innovation for devising integrative analytic methods that relate molecular measurements to clinical phenotypes—particularly by identifying biomarkers for diagnosis, prognosis and personalization of care. This track will focus on systems approaches to integrate multiple kinds of data to facilitate drug discovery, drug repositioning and biomarker discovery.

**TRACK 3: BASE PAIRS TO BEDSIDE**

It is expected that 30,000 people will have their whole genome sequenced just this year. Our current approaches to understand genetic information in the clinical setting need an overhaul. With the increase in the ability to capture phenotypic information – both in the research and clinical settings – our capability to represent and relate phenotypes with genotypes to understand the natural history of diseases has become rate limiting. This track will focus on efforts to link phenotypes in clinical descriptions with the massive amounts of individual genomic information that will be available soon.

**TRACK 4: INFORMATICS WITH BIG DATA**

Given the rapid advances in natural language processing and access to vast computing infrastructure as well as sophisticated ontologies, data-mining and machine learning tools have converged in a manner that allows us to perform “Big Data” analyses. Simultaneously, because of the changes in public policy, information technology, and electronic health record (EHR) adoption, large datasets are increasingly available in healthcare bringing us close to the “threshold of sufficient data.” Web-scale data mining efforts have shown that algorithms behave differently when applied to very large datasets and that with sufficient data simple approaches can work well. This track will focus on efforts that apply Web-scale data-mining methods on massive datasets.

**LEARNING OBJECTIVES**

- To present the latest progress on using informatics approaches to improve translational biomedical research
- To demonstrate how molecular bioinformatics can enhance clinical research, genetic medicine, and healthcare
- To demonstrate how clinical informatics deploying molecular data and knowledge can contribute to the delivery of molecular medicine and individualized health management
- To identify the current challenges of translational bioinformatics, articulate opportunities, and to define the future directions
- To identify areas of interaction among computational biology, genomics research, statistical genetics, electronic health records, health information exchanges, and public health
- To establish a framework for developing, deploying and assessing translational bioinformatics initiatives
- To provide a platform to share research-related issues among the nationwide initiatives on translational research informatics, such as CTSA, NCBC, caBIG, etc.
TREY IDEKER, PhD

Division Chief of Medical Genetics
University of California San Diego School of Medicine

Dr. Ideker is Chief of Genetics at the UCSD School of Medicine. He also serves as Professor of Bioengineering, Adjunct Professor of Computer Science and Member of the Moores UCSD Cancer Center. Ideker received Bachelor’s and Master’s degrees from MIT in Electrical Engineering and Computer Science and his Ph.D. from the University of Washington in Molecular Biology under the supervision of Dr. Leroy Hood. He is a pioneer in using genome-scale measurements to construct network models of cellular processes and disease. His recent research activities include assembly of networks governing the response to DNA damage, development of software for protein network cross-species comparisons, and network-based diagnosis of disease. Ideker serves on the Editorial Boards for Bioinformatics and PLoS Computational Biology, Board of Directors for US-HUPO and the Cytoscape Consortium, and is a regular consultant for companies such as Monsanto, Genstruct, and Mendel Biotechnology. He was named one of the Top 10 Innovators of 2006 by Technology Review magazine and the 2009 Overton Prize recipient from the International Society for Computational Biology. His work has been featured in news outlets such as The Scientist, the San Diego Union Tribune, and Forbes magazine.

HOWARD J. JACOB

Warren P. Knowles Chair of Genetics
Director, Human and Molecular Genetics Center
Professor, Departments of Physiology and Pediatrics
Vice Chair of Research in Pediatrics
Medical College of Wisconsin

Professor Jacob received his Ph.D. in Pharmacology from the University of Iowa in 1989. He completed two parallel post-doctoral fellowships in functional genomics and molecular genetics at Harvard, Stanford and MIT with Victor J. Dzau, M.D. and Eric S. Lander, Ph.D. He was on the faculty at Massachusetts General Hospital and Harvard Medical School for nearly 4 years before moving to Milwaukee. He joined the Medical College of Wisconsin in 1996 as an Associate Professor, Department of Physiology with full Professorship and Tenure in 2001. He was appointed the Founding Director of the Human and Molecular Genetics Center (HMGC) and was awarded the Warren P. Knowles Chair of Genetics in 1999. He has published more than 200 peer reviewed articles.

In 2010 Dr. Jacob led a team of researchers at the Medical College who used an innovative DNA sequencing technique to unravel the medical mystery of Nicholas Volker, a young boy whose life-threatening disease had baffled his doctors and tested his family’s faith. Working with Medical College scientists and physicians at the Children’s Hospital of Wisconsin, Dr. Jacob’s team used Nicholas’ DNA to diagnose his disease and recommend a course of treatment. This treatment has so far been successful.
KEYNOTE SPEAKERS

WEDNESDAY, MARCH 21, 2012

8:30 – 10:00 am

Robert M. Califf, MD
Vice Chancellor for Clinical Research
Professor of Medicine, Division of Cardiology
Duke University’s Translational Medicine Institute

Vice Chancellor for Clinical Research, Director of the Duke Translational Medicine Institute (DTMI), and Professor of Medicine, Cardiology, Duke University Medical Center, Dr. Robert Califf leads a large, multifaceted organization focused on transforming the path for scientific discoveries into improved medical care. Prior to his role at DTMI, he was the founding Director, Duke Clinical Research Institute, an academic research organization. He is the editor-in-chief of American Heart Journal, and has authored more than 1000 peer-reviewed articles.

He has served on the Cardiorenal Advisory Panel of the U.S. Food and Drug Administration and the Pharmaceutical Roundtable of the Institute of Medicine (IOM). He co-chairs the Clinical Trials Transformation Initiative, a public private partnership focused on improving the clinical trials system, as is the Director of the Clinical Research Forum, an organization of academic health and science system leaders working to enhancing the effectiveness of the clinical research enterprise. A member of the Drug Discovery, Development, and Translation Forum of the IOM, Dr. Califf also participates as a member of the National Institute of Health Council on Aging, and President Obama’s Council of Advisors on Science and Technology.

3:30 – 5:00 pm

Russ Altman, MD, PhD
Professor of Bioengineering, Genetics, and Medicine
(and Computer Science, by courtesy)
Chairman, Department of Bioengineering
Director, Biomedical Informatics Training Program
Stanford University

The importance of Translational Bioinformatics continues to grow in biomedical research, genetics, education, and diagnostic and therapeutic discovery. In the past year, we have seen continued progress in linking clinical data to molecular data, as we usher in a new era of molecular medicine. This session will review notable publications over the past twelve months, highlighting trends and milestones achieved in the past year. This is the fifth installment of this keynote talk by Dr. Altman.
**T01: Introduction to Translational Bioinformatics**

*I. Sarkar, University of Vermont; J. Tenenbaum, Duke University*

In 2005, Dr. Elias Zerhouni, Director of the National Institutes of Health (NIH), wrote “It is the responsibility of those of us involved in today’s biomedical research enterprise to translate the remarkable scientific innovations we are witnessing into health gains for the nation... At no other time has the need for a robust, bidirectional information flow between basic and translational scientists been so necessary.” Clearly evident in Dr. Zerhouni’s quote is the role biomedical informatics needs to play in facilitating translational medicine. American Medical Informatics Association (AMIA) now hosts the Joint Summits on Translational Science of which the Summit on Translational Bioinformatics is one of the two components. This tutorial is designed to teach the basics of the various types of molecular data and methodologies currently used in bioinformatics and genomics research, and how these can interface with clinical data. This tutorial will address the hypotheses one can start with by integrating molecular biological data with clinical data, and will show how to implement systems to address these hypotheses. The tutorial will cover real-world case-studies of how genetic, genomics, and proteomic data has been integrated with clinical data.

**T02: Ontology Services for Translational Research in the i2b2 Workbench**

*S. Murphy, Massachusetts General Hospital; R. Ferguson, Stanford University*

The i2b2 platform uses vocabularies extensively in the querying and manipulation of patient data for translational and clinical research. The National Center for Biomedical Ontology offers an extensive range of Web services for accessing ontologies, generating value sets and lexicons, annotating data, and performing information retrieval that form key elements of software systems in informatics. Given the importance of the use of ontologies for data integration, NCBO and i2b2 have initiated a collaboration to provide access to cutting edge ontology services from within the i2b2 workbench to enable cross institution data transformation. This tutorial will review the drivers of this collaboration, provide experience in using the NCBO’s resources, and will offer participants in-depth understanding of how ontologies and terminologies are used in biomedical informatics. This tutorial will review the use of ontologies in i2b2 and discuss their integration with the NCBO’s Ontology Web Services infrastructure.
MONDAY, MARCH 19, 2012  
continued

8:30 am – 12:00 pm  
**Tutorials**

**T03: Reusing EHRs for Clinical, Genomic, and Pharmacogenomic Discovery at Vanderbilt and within the eMERGE Network**  
Room: Stockton  
*J. Denny, H. Xu, Vanderbilt University*

This tutorial will cover basic themes about use of EHRs for generating cohorts of patients to serve as cases and controls for given clinical phenotypes. EHRs can be used for many different types of research include disease-based, response to treatment, clinical biomarkers, redefining “normal”, and analysis of changes over time of clinical variables and parameters. Deriving such phenotypes from EHR data can be challenging. Methods typically involve use of billing data, medication records (often unstructured), laboratory data, and natural language processing. After derivation of these phenotypes, populations can be used for clinical research. Linkage to DNA biobanks also enables the possibility of genomic and pharmacogenomic associations. Research within the eMERGE network has demonstrated success with EHR-based genome-wide association studies (GWAS). In addition, use of EHR-linked genetic data uniquely enables phenome-wide associations studies (PheWAS), which allows an unbiased scan of what diseases may be associated with a given genotype.

**T04: Introduction to R for Bioinformatics and Biomedicine**  
Room: Balboa  
*David Ruau, Stanford University*

Bioconductor, a package repository for bioinformatics, contains 467 packages in addition to the 3128 general-purpose packages from R. The wide array of possibilities make R a platform particularly suited for translational bioinformatics research. However, like other statistical software, the learning curve can be steep for some of us less versed in computer science. This tutorial is based on the successful workshop “Introduction to R Programming” taught at Stanford. Participants will be introduced to the basics of the R language through practical examples from real biomedical research projects. We will show advanced techniques on how different resources can be plugged into R to perform an analysis and produce publication ready graphics.

10:00 – 10:30 am  
Coffee Break  
Room: Cyril Magnin Foyer

1:30 – 3:00 pm  
**TBI Opening Plenary Session and Keynote Address:**  
Room: Cyril Magnin I/II  
*Dr. Trey Ideker*  
University of California San Diego

See page 8 for more information

3:00 – 3:30 pm  
Coffee Break  
Room: Cyril Magnin Foyer
3:30 – 5:00 pm

**Scientific Sessions**

**TBI-01: Papers and Poster Bullets: Predictions from Large Datasets**

Track 1, 4  
Room: Cyril Magnin I  
Session Chair: Shyam Vishwesaran

**Papers**

An Automated Bayesian Framework for Integrative Gene Expression Analysis and Predictive Medicine  
*N. Parikh, MIT; A. Zollanvari, Harvard Medical School; G. Alterovitz, Harvard Medical School*

Large-scale Prediction of Adverse Drug Reactions by Integrating Chemical, Biological, and Phenotypic Properties of Drugs  
*M. Liu, Y. Wu, Y. Chen, J. Sun, Z. Zhao, Vanderbilt University; X. Chen, University of Kansas; H. Xu, Vanderbilt University*

From Sequencer to Supercomputer: An Automatic Pipeline for Managing and Processing Next Generation Sequencing Data  
*T. Camerlengo, H. Ozer, R. Onti-Srinivasan, P. Yan, T. Huang, J. Parvin, K. Huang, The Ohio State University*

**Poster Bullets**

Application of Laboratory Information System to enhance Translation Research  
*W. Amin, H. Singh, University of Pittsburgh; A. Parwani, University of Pittsburgh Medical Center*

The Architecture behind the Identity Management Cell within i2b2  
*M. Mendis, S. Murphy, L. Phillips, R. Kuttan, J. Donahoe, Partners HealthCare; S. Churchill; I. Kohane, Harvard University*

Using Semantic Technologies for Phylogeny  
*M. Panahiazar, Ohio Center of Excellence in Knowledge-Enabled Computing; J. Leebens-Mack, University of Georgia; A. Ranabahu, A. Sheth, Ohio Center of Excellence in Knowledge-Enabled Computing*

The Role of Method Intuition in Translational Ethics  
*S. Bhavnani, J. Farroni, J. Crowder, B. McKinney, R. Pillai; W. Calhoun, R. Rose, M. Carter, University of Texas Medical Branch*
3:30 – 5:00 pm  Scientific Sessions

TBI-02: Podium Abstracts and Poster Bullets: Disease Informatics
Track 1, 2, 3
Room: Cyril Magnin II
Session Chair: Madhavi Ganapathiraju

Podium Abstracts

Expression Blast: Within and Between Species Expression Comparison Tool Applied to Determine Molecular Mechanisms in Aging

Shared Genetic Risk factors between Autism and Autoimmune Disorders
J. Jung, D. Wall, Harvard Medical School

Novel Method for Meta-analysis of Acute Rejection across Multiple Organ Data Sets Identifies a Core Immune Response Module for Repositioning Novel Therapeutics in Transplantation

A Human Developmental Knowledge Base for Disease Informatics
C. Mungall, Lawrence Berkeley National Laboratory; C. Torniai, Oregon Health & Science University; J. Bard, Weatherall Institute of Molecular Medicine; G. Gkoutos, University of Cambridge; S. Essaid, Oregon Health & Science University; P. Schofield, University of Cambridge; P. Robinson, Charité-Universitätsmedizin Berlin; D. Smedley, European Bioinformatics Institute; S. Lewis, Lawrence Berkeley National Laboratory; M. Haendel, Oregon Health & Science University

Poster Bullets

Full-genome Analysis and Prediction of Causal Regulatory Variations
V. Bernard, P. Tan, D. Arenillas, W. Wasserman, Centre for Molecular Medicine and Therapeutics (CMMT)

Genetic Variations Associated with Age-of-Onset of Alzheimer’s disease
A. Wong, University of Pittsburgh; S. Bhavnani, University of Texas Medical Branch; S. Visweswaran, University of Pittsburgh

Translational Bioinformatics Pipeline for Next-Generation Sequencing: Identifying Biomarkers for Clinical Prediction
J. Phan, Georgia Institute of Technology/Emory University; P. Wu, Georgia Institute of Technology; M. Wang, Georgia Institute of Technology/Emory University

Comprehensive Characterization of an Individual’s LINE-1 Insertion Profile using Next Generation Sequencing Data
T. Kalbfleisch, A. Kemper, K. Ramos, M. Hindi, University of Louisville
Scientific Sessions

TBI-03: Podium Abstracts and Poster Bullets: Mining the EHR

Track 1, 3, 4
Room: Mission
Session Chair: Paea LePendu

Podium Abstracts

Interpreting Lagged Linear Correlation and Using Range to Prioritize
G. Hripcsak, D. Albers, A. Perotte, Columbia University

A Personal Risk Prediction Model for Prostate Cancer based on specific Family History Constellation
F. Albright, Pharmacotherapy Outcomes Research Center; R. Stephenson, L. Cannon-Albright, University of Utah School of Medicine

A Translational Bioinformatics Approach to Drug-drug Interaction (DDI) Identification and Validation
X. Han, Indiana University School of Medicine; Z. Wang, A. Subhadarshini, Indiana University; S. Karnik, Indiana University; R. Strother, D. Flockhart, Indiana University School of Medicine; J. Overhage, Regenstrief Institute; S. Quinney, Indiana University; J. Duke, Regenstrief Institute; L. Li, Indiana University School of Medicine

Automated Temporal Causal Inference from EHR Data
S. Kleinberg, G. Hripcsak, Columbia University

Poster Bullets

Asthma Internet Searching: A Surveillance and Rapid Response Opportunity?
R. Divekar, R. Pillai, W. Calhoun, S. Bhavnani, University of Texas Medical Branch

Indexing the UMLS for Large Scale Knowledge Discovery in Biomedical Applications
Y. Xiang, K. Lu, S. James, T. Borlawsky, K. Huang; P. Payne, The Ohio State University

Discovering Latent “Modules of Care” Via Network Analysis Of Electronic Health Records
A. Bauer-Mehren, P. LePendu, S. Iyer, N. Shah, Stanford University

The Pharmacogenomics Knowledge Base (PharmGKB): Bridging the Gap Between Accumulation of Gene-Drug Knowledge and Application in the Clinic
E. McDonagh, Stanford University
Scientific Sessions

TBI-04: Panel: SaaS-Based Translational Research: The Path to Reality for “Research in the Cloud”
Track 1
Room: Cyril Magnin III
Session Chair: Stephen Granite

R. Winslow, The Johns Hopkins University; I. Foster, The University of Chicago; B. Athey, University of Michigan; D. Singh, Amazon Web Services

Like virtually all other areas of biomedical research, translational bioinformatics struggles to evolve with the explosion in available healthcare data. To ride, rather than succumb, to this massive data wave, translational scientists need easy ways to move and mine datasets, without having to become IT experts. Cloud computing promises to alleviate some of these pressures; however, concerns about feasibility still exist for some scientists and their computing facilities, particularly in the areas of security, privacy and compliance. This panel will explore the requirements for a Software as a Service (SaaS) solution that can be used today to transform translational research. Panelists will represent each constituency of a potential online research environment (the clinical researchers, the institutional computing service providers, the data management and transfer service providers, and the cloud computing service providers). The panelists will debate the delta between where we are - and where we need to be - for translational scientists to reliably and securely perform research in the cloud. During this discussion, the panel will address the feasibility concerns through examples of clinical research cloud computing case studies.
8:00 – 9:00 am Coffee and Pastries Room: Cyril Magnin Foyer

8:00 am – 5:00 pm Registration Open Room: Cyril Magnin Foyer

9:00 – 10:30 am Scientific Sessions

TBI-05: Papers and Poster Bullets: Research Platforms
Track 1 Room: Cyril Magnin I
Session Chair: Jessica Tenenbaum

Papers

**A Simulation Platform to Examine Heterogeneity Influence on Treatment**
C. Chi, V. Fusaro, P. Patil, M. Crawford, Harvard Medical School; C. Contant, TIMI Study Group; P. Tonellato, Harvard Medical School

**Towards an Oncology Database (ONCOD) Using a Warehousing Approach**
X. Wang, University of Chicago and Argonne National Laboratory; L. Liu, J. Fackenthal, P. Chang, G. Newsstead, S. Chmura, University of Chicago; I. Foster, University of Chicago and Argonne National Laboratory; O. Olopade, University of Chicago

**The Role of Complementary Bipartite Visual Analytical Representations in the Analysis of SNPs: A Case Study in Ancestral Informative Markers**
S. Bhavnani, University of Texas Medical Branch; G. Bellala, University of Michigan; S. Victor, University of Texas; M. Abbas, V. McMicken, University of Houston Clear Lake; J. Tupa, University of Houston Clear Lake; S. Visweswaran, University of Pittsburgh

Poster Bullets

**RProteomics on the Cancer Bioinformatics Grid (caBIG®)**

**Enhanced Collaboration and Research Discovery through VIVO**
K. Holmes, Washington University School of Medicine; M. Conlon, University of Florida; Y. Ding, Indiana University

**Technical Approach to Guarding Patient Privacy when Recruiting Patient within i2b2**
M. Mendis, S. Murphy, L. Phillips, R. Kuttan, Partners HealthCare; S. Churchill; I. Kohane, Harvard University; J. Donahoe, Partners HealthCare

**Update: CTSA Omics Data Standards Affinity Group**
D. Handler, Northwestern University; A. Lynn, J. Tenenbaum, Duke University
9:00 – 10:30 am  

**Scientific Sessions**

**TBI-06: Podium Abstracts and Poster Bullets: Semantic Querying**  
Track 1, 4  
Session Chair: Mark Musen  
Room: Cyril Magnin II

**Podium Abstracts**

*Supercharging i2b2: Gaining Orders of Magnitude of Performance in a Large Clinical Data Repository*  
G. Weber, Harvard Medical School

*Syntactic-Semantic Question Frames for Cohort Identification*  
D. Demner-Fushman, S. Abhyankar, US National Library of Medicine

*An Informatics Infrastructure for Translating Pharmacogenomic Knowledge into Clinical Practice*  
M. Samwald, Medical University of Vienna; A. Coulet, 3LORIA - INRIA Nancy - Grand-Est; R. Freimuth, Mayo Clinic; I. Huerga, Linkatu, S.L.; J. Luciano, Rensselaer Polytechnic Institute; E. Pichler, W3C; R. Powers, Predictive Medicine, Inc.; E. Prud’hommeaux, World Wide Web Consortium; F. Whipple, Genomics Education Initiative; M. Marshall, Leiden University Medical Center; M. Dumontier, Carleton University

**Semantic Technology and Translational Genomic Research**  
M. Panahiazar, A. Sheth, A. Ranabahu, Ohio Center of Excellence in Knowledge-Enabled Computing; J. Leebens-Mack, University of Georgia

**Poster Bullets**

*The Importance of using the Age-dependent Reference Ranges of the Serum TC obtained from the Cohort Corresponding to the Following up Patients*  
Y. Kurihara, T. Takao, Kochi Medical School; S. Irino, Government of Ehime Prefecture

*Representing Multiple Standards in a Single DAM: use of Atomic Classes*  
S. Mungal, Duke Comprehensive Cancer Center; M. Walker, Mead Walker Consulting; R. Wilgus, Duke Clinical Research Institute; D. Pinchotti, American College of Cardiology; D. Kong, Duke Clinical Research Institute; J. Tcheng, DCRI; W. Barry, Duke Cancer Institute; B. McCourt, Duke Clinical Research Institute

*Using NICU Data to Understand Physiology and Identify Damage in Patients with Acute Brain Injury*  
9:00 – 10:30 am

Scientific Sessions

TBI-07: Papers and Poster Bullets: Methods for Large Datasets
Track 1, 2, 4
Room: Mission
Session Chair: Neil Sarkar

Papers

A Partitioning Based Adaptive Method for Robust Removal of Irrelevant Features from High-dimensional Biomedical Datasets
G. Liu, L. Kong, Pennsylvania State University; V. Gopalakrishnan, University of Pittsburgh

Formal Concept Analysis of Disease Similarity
B. Keller, Eastern Michigan University; F. Eichinger, M. Kretzler, University of Michigan

Stochastic Model Search with Binary Outcomes for Genome-wide Association Studies
A. Russu, G. Botta, A. Malovini, University of Pavia; F. Villa, A. Puca, IRCCS Multimedica; R. Bellazzi, Università di Pavia

Poster Bullets

Pharmacokinetics Numerical Parameter extraction from Article Tables
Z. Wang, Indiana University

Integration of Whole-genome Functional Annotations from Multiple Sources, Applied to Human Pathogens
E. Cadag, A. Huang, A. Navid, C. Zhou, P. D’haeseleer, Lawrence Livermore National Laboratory

Optimal Probeset Filtering Using Proteomic-Transcriptomic Correlations.
K. McDade, R. Day, U. Chandran, A. Lisovich, University of Pittsburgh

Using Plasma Proteomic Analysis for Venous Thromboembolism Risk Stratification in Cancer Patients
H. Itakura, A. Holzer, L. Hofmann, P. Tsao, Stanford University

TBI-08: Panel: Practical experience with linked open biomedical data: State of the Art and Future Directions
Track 1
Room: Cyril Magnin III
D. Bourges-Waldegg, Harvard University; C. Barnes, University of Florida; A. Ruttenberg, University of Buffalo; Y. Ding, Indiana University; C. Torniai, Oregon Health & Science University

A large portion of the valuable data produced in biomedical research remains inaccessible to the research community. In recent years, Semantic Web technologies and the Linked Open Data initiative have gained traction as the gold standard for integrating, linking, and sharing biomedical data. The technology of the Semantic Web has advanced considerably, however its potential and adoption have yet to be fully realized. There still exist challenges related to Linked Open Data’s consistency of semantic representation, scalability, provenance, and privacy. This panel includes representatives of the eagle-i, VIVO, NeuroCommons, and Chem2BioRDF projects, all of which aim to build semantic representations of biomedical information and publish it as Linked Data. In order to maximize interoperability and value of the resulting data sets, each of these projects has reused existing ontologies and referenced other data sets where possible. The panelists will present their experience and engage the audience on topics including i) Producing, maintaining, and consuming Linked Biomedical Data; ii) Lessons learned in developing frameworks for large-scale sharing of biological information on the Web; iii) Using clinical data sources in the context of Linked Data.
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<td>10:30 – 11:00 am</td>
<td>Coffee Break</td>
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<td>11:00 am – 12:00 pm</td>
<td><strong>Keynote Presentation</strong></td>
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<td>12:15 – 1:15 pm</td>
<td><strong>Lunch &amp; Learn</strong></td>
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**Scientific Sessions**

**TBI-10: Podium Abstracts and Poster Bullets: Data Mining**

Track 1, 3, 4  
Room: Cyril Magnin II

Session Chair: Sean Mooney

**Podium Abstracts**

Hyper-experiments: Bayesian Inference and Annotation for GEO  
*K. Marwah, MIT; A. Zollanvari, G. Alterovitz, Harvard Medical School*

Prediction-based Bayesian Network Analysis of Gene Sets for Genome-wide Association and Expression Studies  
*A. Zollanvari, Harvard Medical School*

Identification of Potential Model Organisms for Disease Using A Vector Space Model Approach  
*I. Sarkar, University of Vermont*

Replicating Genetic Associations in Small, Machine Learning Defined Cohorts  
*R. Carroll, A. Eyler, R. Zink, J. Denny, Vanderbilt School of Medicine*

**Poster Bullets**

A Semantic Proteomics Dashboard (SemPoD) for Proteomics Data Management in Translational Research  
*C. Jayapandian, R. Ewing, S. Sahoo, G. Zhang, Case Western Reserve University*

Personalizing the Return of Genome Sequencing Results: Consumer Health Informatics Meets Bioinformatics  
*N. Anderson, A. Hartzler, H. Tabor, M. Bamshad, University of Washington*

Integrated Mobile Health Tools for Monitoring of Treatment and Transition of Adolescents with Sickle Cell Disease  
*T. Stokes, Emory University & Georgia Tech; R. Brown, Emory University, Department of Pediatrics; C. Cheng, Georgia Institute of Technology; C. Dampier, T. New, Emory University; M. Wang, Georgia Institute of Technology/Emory University*

A Patient-entered Family Health History Tool Using Clinical Decision Support (CDS) to Generate Personalized Risk Assessment Messages During the First Prenatal Care Visit  
*B. Lin, March of Dimes; E. Edelman, National Coalition Health Professional Education in Genetics; T. Doksum, Toksum Consulting; K. Hughes, Massachusetts General Hospital; B. Drohan, Massachusetts General Hospital; S. Dolan, Albert Einstein College of Medicine; V. Edelson, J. O’Leary, Genetic Alliance; A. Harris, S. Copeland, Health Resources and Services Administration; J. Scott, National Coalition Health Professional Education in Genetics*
1:30 – 3:00 pm  

Scientific Sessions

TBI-11: Podium Abstracts and Poster Bullets: Tools and Communities
Track 1, 2, 3, 4  
Room: Mission
Session Chair: David Ruau

Podium Abstracts

A Community Approach for Improving Bioinformatics Programming Skills
P. Fearn, P. Tarczy-Hornoch, University of Washington

The MURDOCK Study: A Framingham for the Genomic Era

Bioinformatic Data Integration to Assess Data Preprocessing

Special Presentation - Integrative Computational Strategies for Drug Safety: EU-ADR and eTOX projects
D. Heard, Novartis Institutes for Biomedical Research; F. Sanz, Fabra University

Poster Bullets

A Prediction-based Bayesian Framework for Quantifying the Interaction of Demographics and Genetics: Application to Alcohol Dependence
A. Zollanvari, Harvard Medical School; J. Thomas, MIT; G. Alterovitz, Harvard University

Multinet Bayesian Networks for Integrative Genomic Discovery: Application to Genetic Epistatic Interactions in HIV
A. Zollanvari, Harvard Medical School; A. Wu, MIT; G. Alterovitz, Harvard

Integrative Bioinformatics Approach to Candidate Pain Gene Identification
D. Ruau, J. Dudley, R. Chen, L. Lazzeroni, D. Clark, M. Angst, Stanford University School of Medicine; A. Butte, Stanford University

Towards Time Modeling of Vaccine Adverse Events
C. Tao, Mayo Clinic; Y. He, University of Michigan Medical School
Scientific Sessions

TBI-12: Panel: Knowledge Synthesis for in silico Science: Lessons Learned and Future Directions
Track 1
Room: Cyril Magnin III

P. Payne, The Ohio State University; I. Sarkar, University of Vermont; P. Tarczy-Hornoch, University of Washington; P. Tonellato, Harvard Medical School

Given an increasing focus throughout the biomedical community on clinical and translational research paradigms, there has been a corresponding and significant growth in data and knowledge collections available for primary and secondary interrogation by investigators. As the availability of such resources increases, the ability to reason upon networks of biological and molecular markers, phenotypic variables, and available data sets, in order to synthesize knowledge related to biologically or clinically relevant interrelationships between such entities is highly desirable. Numerous techniques exist that can be used to induce networks of co-expressed or otherwise related features within homogenous data sets and information constructs. However, the contemporary biomedical informatics knowledge base is noticeably lacking reports of integrative approaches that bridge such methodological approaches and place their results in actionable contexts in order to enable knowledge synthesis. This panel will explore the state-of-the-art relative to evolving in silico knowledge synthesis techniques, intended to address the preceding gap in knowledge and practice. In addition, panelists will discuss future directions for methodological innovation in this emergent and critical domain.

3:00 – 3:30 pm Coffee Break Room: Cyril Magnin Foyer

Scientific Sessions

TBI-13: Papers and Poster Bullets: Data Integration Methods
Track 1, 3 4 Room: Cyril Magnin I

Session Chair: Gil Alterovitz

Papers

A Selective Voting Convex-Hull Ensemble Procedure for Personalized Medicine
R. Nagarajan, University of Arkansas for Medical Sciences; R. Kodell, University of Arkansas

Context-Specific Ontology Integration: A Bayesian Approach
K. Marwah, D. Katzin, MIT; A. Zollanvari, Harvard Medical School; N. Noy, Stanford University; M. Ramoni, G. Alterovitz, Harvard Medical School

Incorporating Molecular and Functional Context into the Analysis and Prioritization of Human Variants Associated with Cancer
T. Peterson, N. Nehrt, D. Park, M. Kann, University of Maryland, Baltimore County

Therapy Targeted at Unique Personal Disease Polymorphism Using Mechanistic Concordance between Mendelian and Complex Inheritance Anchored on Protein Domain Interactions
K. Regan, University of Illinois at Chicago/University of Chicago; K. Wang, University of Chicago; E. Doughty, University of Maryland, Baltimore County; H. Li, J. Li, University of Illinois at Chicago/University of Chicago; Y. Lee, University of Chicago; M. Kann, University of Maryland, Baltimore County; Y. Lussier, University of Illinois at Chicago/University of Chicago

Poster Bullets

An online Platform for Integration and Sharing of Phenotypic and Genomics Information
E. Portales-Casamar, F. Lui, N. St-Georges, A. Zoubarev, P. Pavlidis, University of British Columbia

Genome Analysis and Clinical Translation for Patients with Intellectual Disability
C. Shyr, UBC; V. Bernard, D. Arenillas, Centre for Molecular Medicine and Therapeutics (CMMT); S. Stockler, C. van Karnebeek, UBC; W. Wasserman, Centre for Molecular Medicine and Therapeutics (CMMT)

Representation of Radiology Differential Diagnoses using OWL
C. Kahn, Jr., J. Budovec, C. Lam, S. Goth, Medical College of Wisconsin
3:30 – 5:00 pm

Scientific Sessions

TBI-14: Podium Abstracts and Poster Bullets: Analyses with Phenotypes
Track 1  
Room: Cyril Magnin II
Session Chair: Anna Bauer-Mehren

Podium Abstracts

Systematic Drug Repositioning based on Clinical Side-effects (DRoSef)
L. Yang, P. Agarwal, GlaxoSmithKline

A Multi-scale Neurodegenerative Disease-phenotype Network
C. Mungall, Lawrence Berkeley National Laboratory; S. Maynard, F. Imam, University of California, San Diego; S. Lewis, Lawrence Berkeley National Laboratory; M. Martone, University of California San Diego

Integrative Decision-making Tools to Support Regenerative Medicine
F. Mulas, University of Pavia; L. Zagar, University of Ljubljana; B. Zupan, University of Ljubljana; R. Bellazzi, Università di Pavia

Poster Bullets

Automated Coding of Key Cancer Concepts from Text Based Electronic Pathology Reports
E. Durbin, T. Gal, University of Kentucky; G. Cernile, Artificial Intelligence in Medicine, Inc.; T. Johnson, University of Kentucky

An Integrated Pharmacokinetics Ontology and Semantically Annotated Corpus for Text Mining
A. Subhadarshini, Indiana University Purdue University Indianapolis; S. Karnik, Z. Wang; S. Philips, Indiana University; J. Duke, Regenstrief Institute, Inc.; S. Quinney, D. Flockhart, L. Li, X. Han, Indiana University

Extraction of in-vivo Pharmacokinetic Drug-drug Interactions from Biomedical Literature Using Pattern Based and Machine Learning Methods
S. Karnik, A. Subhadarshini, Z. Wang, X. Han, S. Quinney, L. Li, Indiana University

A Framework to Search and Extract Pharmacokinetics Parameter Numerical Data from Literatures
Z. Wang, Indiana University

TBI-15: Panel: Detection and Prediction of Adverse Drug Events in Clinical and Molecular Data
Track 3  
Room: Cyril Magnin III

N. Tatonetti, R. Altman, Stanford University; I. Kohane, Harvard Medical School; A. Butte, Stanford University; C. Friedman, Columbia University; N. Shah, Stanford University

Adverse drug events (ADEs) are a leading cause of morbidity and mortality around the world, accounting for 100,000 deaths each year. Many ADEs are not observed in clinical trials and their occurrence is unexpected. Organizations, such as the Food and Drug Administration (FDA), have set up adverse event reporting systems to monitor the long term safety of drugs after they hit the market. The increased adoption of electronic medical record (EMR) systems represents another opportunity to study and monitor ADEs at the population level. High-throughput databases of molecular drug assays represent yet another opportunity to detect and predict adverse events. In this panel we explore how different systems and analytical methods are used to identify adverse drug events. In particular, we will focus on the cardiovascular effects of rofecoxib. An association that has been retrospectively replicated using a variety of distinct methodologies. We will explore the challenges of using large clinical data repositories for ADE discovery as well as the methodological innovations that will be necessary to address these challenges.
3:30 – 5:00 pm

Scientific Sessions

TBI-16: Panel: tranSMART: an Open Source Analytical and Data Sharing Informatics Platform Enabling Translational Research

Track 1

B. Athey, K. Smith, University of Michigan

tranSMART is an informatics platform developed by Johnson & Johnson (J&J) that has been placed in open source that enables translation research. The University of Michigan and key academic and industry partners are developing a next-generation analytical and data sharing informatics platform based on tranSMART 1.0. The platform will accelerate biomedical discovery by enabling collaboration among clinicians, researchers and informatics professionals, and will be sustained through an academic, government, and industry private-public partnership. This platform will coalesce and build upon NIH-funded informatics initiatives such as the NCBCs, national data sharing and policy mandates, and leverage existing and emerging national and international research networks. Panel members will provide 1) a technical overview of the tranSMART architecture, 2) an overview of representative early adopters in the areas of cancer (Sage Bionetworks), neuroscience (One Mind for Research) and European plans to develop a knowledge management framework based on tranSMART, 3) an update on efforts to establish a sustainable ecosystem for consensus community-building, platform support and ongoing development, as well as 4) three-year vision for enhancing tranSMART that leverages NIH-funded projects (NCIBI, i2b2, NCBO), and emerging public-private partnerships that scales locally, regionally and nationally.

5:00 – 6:00 pm

Poster Session and Reception

Room: Cyril Magnin Foyer

Using NICU Data to Understand Physiology and Identify Damage in Patients with Acute Brain Injury (Board 1)


Application of Laboratory Information System to Enhance Translation Research (Board 2)

W. Amin, H. Singh, University of Pittsburgh; A. Parwani, University of Pittsburgh Medical Center

Personalizing the Return of Genome Sequencing Results: Consumer Health Informatics Meets Bioinformatics (Board 3)

N. Anderson, A. Hartzler, H. Tabor, M. Bamshad, University of Washington

Discovering Latent “Modules of Care” Via Network Analysis of Electronic Health Records (Board 4)

A. Bauer-Mehren, P. LePendu, S. Iyer, N. Shah, Stanford University

Full-genome Analysis and Prediction of Causal Regulatory Variations (Board 5)

V. Bernard, P. Tan, D. Arenillas, W. Wasserman, Centre for Molecular Medicine and Therapeutics (CMMT)

The Role of Method Intuition in Translational Ethics (Board 6)

S. Bhavnani, J. Farroni, J. Crowder, B. McKinney, R. Pillai, W. Calhoun, R. Rose, M. Carter, University of Texas Medical Branch

Integration of Whole-genome Functional Annotations from Multiple Sources, Applied to Human Pathogens (Board 7)

E. Cadag, A. Huang, A. Navid, C. Zhou, P. D’haeseleer, Lawrence Livermore National Laboratory

Asthma Internet Searching: A Surveillance and Rapid Response Opportunity? (Board 8)

R. Divekar, R. Pillai, W. Calhoun, S. Bhavnani, University of Texas Medical Branch
5:00 – 6:00 pm  
**Poster Session and Reception**  
Room: Cyril Magnin Foyer

- **Automated Coding of Key Cancer Concepts from Text Based Electronic Pathology Reports (Board 9)**  
  E. Durbin, T. Gal, University of Kentucky; G. Cernile, Artificial Intelligence in Medicine, Inc.; T. Johnson, University of Kentucky

- **Integration of Phenotype Data Using Multiple Ontologies for Disease Model Identification (Board 10)**  

- **Update: CTSA Omics Data Standards Affinity Group (Board 11)**  
  D. Handler, Northwestern University; A. Lynn, J. Tenenbaum, Duke University

- **Enhanced Collaboration and Research Discovery through VIVO (Board 12)**  
  K. Holmes, Washington University School of Medicine; M. Conlon, University of Florida; Y. Ding, Indiana University

- **IDR Snapshot: Quantitative Assessment Methodology evaluating Size and Comprehensiveness of an Integrated Data Repository (Board 13)**  
  V. Huser, National Institutes of Health, Clinical Center; J. Cimino, NIH

- **Using Plasma Proteomic Analysis for Venous Thromboembolism Risk Stratification in Cancer Patients (Board 14)**  
  H. Itakura, A. Holzer, L. Hofmann, P. Tsao, Stanford University

- **A Semantic Proteomics Dashboard (SemPoD) for Proteomics Data Management in Translational Research (Board 15)**  
  C. Jayapandian, R. Ewing, S. Sahoo, G. Zhang, Case Western Reserve University

- **Representation of Radiology Differential Diagnoses using OWL (Board 16)**  
  C. Kahn, Jr., J. Budovec, C. Lam, S. Goth, Medical College of Wisconsin

- **Comprehensive Characterization of an Individual’s LINE-1 Insertion Profile using Next Generation Sequencing Data (Board 17)**  
  T. Kalbfleisch, A. Kemper, K. Ramos, M. Hindi, University of Louisville

- **Extraction of in-vivo Pharmacokinetic Drug-drug Interactions from Biomedical Literature Using Pattern Based and Machine Learning Methods (Board 18)**  
  S. Karnik, A. Subhadarshini, Z. WangX. Han, S. Quinney, L. Li, Indiana University

- **The Importance of Using the Age-dependent Reference Ranges of the Serum TC obtained from the Cohort Corresponding to the Following up Patients (Board 19)**  
  Y. Kurihara, T. Takao, Kochi Medical School; S. Irino, Government of Ehime Prefecture

- **A Patient-entered Family Health History Tool using Clinical Decision Support (CDS) to Generate Personalized Risk Assessment Messages During the First Prenatal Care Visit (Board 20)**  
  B. Lin, March of Dimes; E. Edelman, National Coalition Health Professional Education in Genetics; T. Doksum, Toksum Consulting; K. Hughes, B. Drohan, Massachusetts General Hospital; S. Dolan, Albert Einstein College of Medicine; V. Edelson, Genetic Alliance; J. O’Leary, Genetic Alliance; A. Harris, S. Copeland, Health Resources and Services Administration; J. Scott, National Coalition Health Professional Education in Genetics

- **Optimal Probeset Filtering Using Proteomic-transcriptomic Correlations (Board 21)**  
  K. McDade, R. Day, U. Chandran, A. Lisovich, University of Pittsburgh

- **The Pharmacogenomics Knowledge Base (PharmGKB): Bridging the Gap Between Accumulation of Gene-drug Knowledge and Application in the Clinic (Board 22)**  
  E. McDonagh, Stanford University
5:00 – 6:00 pm  

**Poster Session and Reception**  

Room: Cyril Magnin Foyer

**Finding Functional Signatures for Immune Cell Types Via Gene Expression Analysis (Board 23)**  
T. Meehan, The Jackson Laboratory; C. Mungall, Lawrence Berkeley National Laboratory; N. Vasilevsky, M. Haendel, Oregon Health & Science University; J. Blake, The Jackson Laboratory; A. Diehl, University at Buffalo, State University of New York

**The Architecture behind the Identity Management Cell within i2b2 (Board 24)**  
M. Mendis, S. Murphy, L. Phillips, R. Kuttan, J. Donahoe, Partners HealthCare; S. Churchill; I. Kohane, Harvard University

**Technical Approach to Guarding Patient Privacy when Recruiting Patient within i2b2 (Board 25)**  
M. Mendis, S. Murphy, L. Phillips, R. Kuttan, Partners HealthCare; S. Churchill; I. Kohane, Harvard University; J. Donahoe, Partners HealthCare

**RProteomics on the Cancer Bioinformatics Grid (caBIG®) (Board 26)**  

**Representing Multiple Standards in a Single DAM: use of Atomic Classes (Board 27)**  
S. Mungal, Duke Comprehensive Cancer Center; M. Walker, Mead Walker Consulting; R. Wilgus, Duke Clinical Research Institute; D. Pinchotti, American College of Cardiology; D. Kong, J. Tcheng, Duke Clinical Research Institute; W. Barry, Duke Cancer Institute; B. McCourt, Duke Clinical Research Institute

**Using Semantic Technologies for Phylogeny (Board 28)**  
M. Panahiazar, Ohio Center of Excellence in Knowledge-Enabled Computing; J. Leebens-Mack, University of Georgia; A. Ranabahu, A. Sheth, Ohio Center of Excellence in Knowledge-Enabled Computing

**Translational Bioinformatics Pipeline for Next-generation Sequencing: Identifying Biomarkers for Clinical Prediction (Board 29)**  
J. Phan, Georgia Institute of Technology/Emory University; P. Wu, Georgia Institute of Technology; M. Wang, Georgia Institute of Technology/Emory University

**An Online Platform for Integration and Sharing of Phenotypic and Genomics Information (Board 30)**  
E. Portales-Casamar, F. Lui, N. St-Georges, A. Zoubarev, P. Pavlidis, University of British Columbia

**Integrative Bioinformatics Approach to Candidate Pain Gene Identification (Board 31)**  
D. Ruau, J. Dudley, R. Chen, L. Lazzeroni, Stanford University; D. Clark, M. Angst, Stanford University School of Medicine; A. Butte, Stanford University

**Genome Analysis and Clinical Translation for Patients with Intellectual Disability (Board 32)**  
C. Shyr, UBC; V. Bernard, D. Arenillas, Centre for Molecular Medicine and Therapeutics (CMMT); S. Stockler, C. van Karnebeek, UBC; W. Wasserman, Centre for Molecular Medicine and Therapeutics (CMMT)
5:00 – 6:00 pm  

**Poster Session and Reception**  
Room: Cyril Magnin Foyer

**Integrated Mobile Health Tools for Monitoring of Treatment and Transition of Adolescents with Sickle Cell Disease (Board 33)**  
T. Stokes, Emory University & Georgia Tech; R. Brown, Emory University, Department of Pediatrics; C. Cheng, Georgia Institute of Technology; C. Dampier, T. New, Emory University; M. Wang, Georgia Institute of Technology/Emory University

**An Integrated Pharmacokinetics Ontology and Semantically Annotated Corpus for Text Mining (Board 34)**  
A. Subhadarshini, Indiana University Purdue University Indianapolis; S. Karnik, Z. Wang, S. Philips, Indiana University; J. Duke, Regenstrief Institute, Inc.; S. Quinney, D. Flockhart, L. Li, X. Han, Indiana University

**The i2b2 with Japanese Clinical Patients’ Data and miRNA Expression Profiles (Board 35)**  
T. Takai, A. Hoshi, Tokyo Medical and Dental University; R. Akasaka, NEC Co.; Y. Fukuoka, H. Tanaka, Tokyo Medical and Dental University

**Towards Time Modeling of Vaccine Adverse Events (Board 36)**  
C. Tao, Mayo Clinic; Y. He, University of Michigan Medical School

**Pharmacokinetics Numerical Parameter Extraction from Article Tables (Board 37)**  
Z. Wang, Indiana University

**A Framework to Search and Extract Pharmacokinetics Parameter Numerical Data from Literatures (Board 38)**  
Z. Wang, Indiana University

**Genetic Variations Associated with Age-of-Onset of Alzheimer’s Disease (Board 39)**  
A. Wong, University of Pittsburgh; S. Bhavnani, University of Texas Medical Branch; S. Visweswaran, University of Pittsburgh

**Indexing the UMLS for Large Scale Knowledge Discovery in Biomedical Applications (Board 40)**  
Y. Xiang, K. Lu, S. James, T. Borlawsky, K. Huang, P. Payne, The Ohio State University

**A Prediction-based Bayesian Framework for Quantifying the Interaction of Demographics and Genetics: Application to Alcohol Dependence (Board 41)**  
A. Zollanvari, Harvard Medical School; J. Thomas, MIT; G. Alterovitz, MIT, Harvard

**Multinets Bayesian Networks for Integrative Genomic Discovery: Application to Genetic Epistatic Interactions in HIV (Board 42)**  
A. Zollanvari, Harvard Medical School; A. Wu, MIT; G. Alterovitz, Harvard
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<th>Time</th>
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<tr>
<td>7:30 – 8:30 am</td>
<td>Coffee and Pastries</td>
<td>Cyril Magnin Foyer</td>
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<td>7:30 am – 5:00 pm</td>
<td>Registration Open</td>
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<td>8:30 – 10:00 am</td>
<td><strong>CRI Opening Plenary Session and Keynote Presentation</strong></td>
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<td><strong>Dr. Robert M. Califf</strong></td>
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<td>10:00 – 10:30 am</td>
<td>Coffee Break</td>
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<td>10:30 am – 12:00 pm</td>
<td><strong>Scientific Sessions</strong></td>
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<td><strong>TBI-17: Papers: Integrative Genomics</strong></td>
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<td><strong>Session Chair: Peter Tonellato</strong></td>
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<td><strong>Integrative Analysis of Common Neurodegenerative Diseases Using Gene Association, Interaction Network, and mRNA Expression Data</strong></td>
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<td><strong>Y. Liu, M. Koyut, S. Maxwell, Case Western Reserve University; Z. Zhao, Vanderbilt University; M. Chance, Case Western Reserve University</strong></td>
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<td><strong>Identifying Common Genes and Networks in Multi-Organ Fibrosis</strong></td>
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<td><strong>K. Wenzke, The Ohio State University College of Medicine; C. Cantemir-Stone, Davis HLRI; J. Zhang, The Ohio State University; C. Marsh, OSU Medical Center; K. Huang, The Ohio State University</strong></td>
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<td><strong>Integrating Genome and Functional Genomics Data to Reveal Perturbed Signaling Pathways in Ovarian Cancers</strong></td>
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<td><strong>S. Lu, X. Lu, University of Pittsburgh</strong></td>
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<td><strong>Coanalysis of GWAS with eQTLs Reveals Disease-tissue Associations</strong></td>
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<td><strong>H. Kang, A. Morgan, R. Chen, Stanford University; E. Schadt, Mount Sinai School of Medicine; A. Butte, Stanford University</strong></td>
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<td><strong>TBI-18: Panel: Crossing the Omic Chasm: Integrating Omic Data into the EHR</strong></td>
<td>Mission</td>
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<td><strong>Track 3</strong></td>
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<td><strong>J. Starren, Northwestern University; E. Bottinger, Mount Sinai School of Medicine; M. Dente, GE Healthcare; G. Wood, Intermountain Healthcare; J. Hoffman, St. Jude Children’s Research Hospital</strong></td>
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Implicit in the vision for Personalized Genomic Medicine is that Genomic and other Omic (e.g., proteomic, metabolomics, etc.) data will need to be integrated into the Electronic Health Records (EHRs) of the future. Omic data breaks the conventional EHR paradigm in a number of ways. The greatest paradigm shift is that next-generation sequencing allows the collection of large amounts of data about a patient, before the clinical significance of that data is known. Another shift is that the raw data are essentially uninterpretable without the aid of computer analysis, even for a domain expert. A number of groups, including the The Electronic Medical Records and Genomics (eMERGE) Network, the Pharmacogenomics Research Network (PGRN) and the HL7 Clinical Genomics Workgroup, as well as individual EHR vendors, are tackling these challenges. The panel will present the perspective and approaches of the various groups.
10:30 am – 12:00 pm  Scientific Sessions

**TBI-19: Late Breaking Session - National Center for Advancing Translational Sciences**  
Track 3  
Room: Cyril Magnin II  

*S. Friend, Sage Bionetworks; E. Collier, NCATS; B. Munos, InnoThink Center for Research in Biomedical Innovation; V. Hale, Medicines360/OneWorld Health; J. Tenenbaum, Duke University*

With the passage of a fiscal year 2012 spending bill, the National Institutes of Health created the National Center for Advancing Translational Sciences (NCATS), with the stated mission to catalyze the generation of innovative methods and technologies that will enhance the development, testing, and implementation of diagnostics and therapeutics across a wide range of human diseases and conditions. This panel will address the newly created NCATS from a variety of perspectives including the NIH, non-profit research organizations, and pharmaceutical companies. Topics for discussion include:

- Informatics and NCRR functions beyond diagnostics and therapeutic development
- Expanding the precompetitive space
- Rare/neglected diseases
- Partnership with industry

Through presentations and discussion, we hope to address what the new center means for the informatics community, and to identify opportunities to contribute to the NCATS mission from the informatics perspective.

**CRI-01: Panel: Identifying Patients for Clinical Studies from Electronic Health Records: The TREC Medical Records Track**  
Track 1  
Room: Cyril Magnin III  

*W. Hersh, Oregon Health & Science University; E. Voorhees, NIST; I. Provalov, Cengage Learning; D. Demner-Fushman, NLM*

The growing investment in electronic health record (EHR) systems provides an opportunity to augment clinical research by allowing the “secondary use” of clinical data. However, there are a number of challenges to using EHR data, the most significant of which is it being inaccessible and difficult to structure due to it being in mostly free-text form. A related challenge has been a lack of robust collections of such data to facilitate research in this area. This has been addressed by the Text Retrieval Conference (TREC), an annual challenge evaluation for information retrieval (IR) researchers. TREC is usually organized into 5-6 tracks focused on different IR problems. In 2011, TREC instituted a Medical Records Track, focused on the retrieval of medical records that could enable the selection of patients for possible participation in clinical studies. This panel reports on the development and implementation of the track including presentations by two of the track’s research participants.

12:15 – 1:15 pm  Lunch & Learn  

**New Clinical Research Tools: Introducing a Solution to Your Volunteer Recruitment Needs While Maintaining Compliance and Ethical Standards**  
Commercial support provided by Velos
1:30 – 3:00 pm

**Scientific Sessions**

**TBI-20: Panel: Big Data**

Track 4  
Room: Cyril Magnin III  
Session Chair: Nigam Shah

*L. D’Avolio, VA Boston Healthcare System; P. Tonellato, Harvard Medical School; J. Hammerbacher, Cloudera, Inc.; D. Anderson, OptumInsight*

It is well known that we are in the midst of a data deluge. When handling Big Data, it is necessary to recognize that data can be “big” in terms of size (e.g. next gen sequencing data) or can be big in terms of the number of individuals on which there is some data collected (e.g. administrative and claims data in healthcare). Increasingly we are moving towards generating data that is big along both these dimensions. This panel will explore the approaches to store, analyze and interpret Big Data.

**CRI-02: Papers and Papers and Poster Bullets: Natural Language Processing Methods for CRI**

Track 1, 2, 4  
Room: Cyril Magnin I  
Session Chair: Hongfang Liu

**Papers**

Dependency Parser-based Negation Detection in Clinical Narratives  
*S. Sohn, S. Wu, C. Chute, Mayo Clinic*

Semantator: Annotating Clinical Narratives with Semantic Web Ontologies  
*D. Song, Lehigh University; C. Chute, C. Tao, Mayo Clinic*

Feasibility of Pooling Annotated Corpora for Clinical Concept Extraction  
*K. Wagholikar, Mayo Clinic; M. Torii, Georgetown University; S. Jonnalagadda, H. Liu, Mayo Clinic*

**Poster Bullets**

An NLP Framework for Classifying Contraception Use among US Women Veterans  
*M. Scotch, Arizona State University; J. Womack, VHA Connecticut; S. Leung, VA Palo Alto Healthcare System; C. Brandt, Yale University*

An Evaluation of the Informativeness of De-identified Documents  
*J. Friedlin, Regenstrief; B. South, VA Salt Lake City Health Care; S. Shen, O. Ferrandez, University of Utah; N. Nokes, M. Maw, VA Salt Lake City Health Care; M. Samore, S. Meystre, University of Utah*

Prevalence Estimates of Clinical Eponyms in De-Identified Clinical Documents  
*B. South, S. Shen, M. Maw, VA Salt Lake City Health Care; O. Ferrandez, University of Utah; J. Friedlin, Regenstrief; S. Meystre, University of Utah*

Modeling UIMA Type System using Web Ontology Language - towards Interoperability among UIMA NLP Tools  
*H. Liu, Mayo Clinic College of Medicine; M. Torii, Georgetown University; S. Wu, Mayo Clinic; V. Kaggal, Mayo Clinic College of Medicine; C. Chute, Mayo Clinic*
1:30 – 3:00 pm  Scientific Sessions

**TBI-21/CRI-03: ACMI Bridge Day Panel: Informatics Challenges and Solutions for Capturing, Taming, and Analyzing the Data Tsunami: Report of the ACMI 2012 Winter Symposium**

Room: Cyril Magnin II

*S. Bakken, Columbia University; V. Florance, National Library of Medicine; W. Chapman, USCD*

As we move from clinical data warehouses and biobanks to broader conceptualizations of comprehensive health determinants data, numerous challenges arise. To address these challenges, the ACMI 2012 Winter Symposium focused on relevant topics including: 1) database structures and ontologies that support linkages among disparate data types; 2) de-identification strategies: where and how; query tools for extraction of analytical data sets; 3) visualization tools that provide cognitive support for investigators, and 4) sharing across databanks for discovery and translation. Panelists will provide a summary of the symposium and key recommendations from Symposium participants for solutions that address the informatics challenges related to capturing, taming, and analyzing large volumes of heterogeneous data.


Track 3

Room: Mission

*A. Butte, Stanford University; W. Hogarth, Sequoia Capital; K. Ku, N. Shah, Stanford University*

To realize the full benefit of translational bioinformatics, the scientific breakthroughs made in research laboratories need to be translated into commercially viable ventures. This panel will explore the milestones and challenges on that path as well as point out areas where there is market need but not enough research. The panel will also discuss the key steps in commercializing discoveries by means of discussing use cases from the panelists experience.

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<td>Coffee Break</td>
<td>Cyril Magnin Foyer</td>
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<tr>
<td>3:30 – 5:00 pm</td>
<td><strong>TBI Closing Session: Translational Bioinformatics Year in Review</strong></td>
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<td><strong>Dr. Russ Altman</strong></td>
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<td>5:00 – 6:00 pm</td>
<td><strong>Joint Summits Reception</strong></td>
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