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MARCH 18 – 20

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Fellow TBI Summit Attendees and AMIA members,

On behalf of AMIA, the Track Chairs, and the whole Scientific Program Committee, it is my pleasure to welcome you to the 2013 Summit on Translational Bioinformatics (TBI). Since 2008, the TBI Summit has been the premier forum for interacting with leaders in informatics at the interface of biology and health care, offering not only world class science, but also a fantastic networking opportunity. This year we have some exciting new additions to the program, designed to take full advantage of the face-to-face interactions that a conference provides, and that even the most technologically advanced journals cannot.

- In addition to scientific papers and podium presentations on the cutting edge of TBI, this year’s summit features exciting panels and sessions on critical related topics such as governance, ethics, training, and career development.
- We are particularly delighted to present keynotes by Stephen Friend, research visionary and the Founder and CEO of Sage Bionetworks, and Debbie Nickerson of the University of Washington, who has been a pioneer in genomics for more than two decades.
- In addition to the ever-popular Introduction to TBI, this year features Russ Altman on How to Write a Grant, Nigam Shah on Ontologies for Data Driven Medicine, an Introduction to CRI (Clinical Research Informatics), and an interactive mock grant review.
- And finally, selected poster submissions will be featured and discussed by senior researchers in the field (many of whom proudly sport their AMIA “Silver Medallions”) in a special poster highlights session on the first day.

For me personally, AMIA has been a critical part of my professional life. Many of the exciting opportunities I have had, including the opportunity to help organize this conference, I can trace back to the relationships I have formed through AMIA, and the Summits in particular. At the same time, I know firsthand that AMIA can feel somewhat... “clubby” to those who are new to the organization, particularly to those of us who focus on the bio- end of the informatics spectrum.

At only about one quarter the size of the Annual Symposium, I find the Summits to be the perfect size for networking with colleagues old and new. Seasoned researchers can often be counted on to introduce a student or young investigator to their esteemed peers. In that spirit, I would like to challenge every attendee, students in particular, to reach out to someone new during the event. Take a chance, say hello, ask someone about their research. That conversation is likely to be just as valuable as many of the talks or poster sessions you attend—maybe even more so.

In my experience these last 5 years, the biggest problem with TBI is that there is always more than one session of interest going on at a given time. Fortunately, there’s Twitter. Do share your experience and observations with your colleagues—#TBI13. We’ll be watching those tweets, and contributions will not go unnoticed. Finally, feel free to contact me via email jessie.tenenbaum@duke.edu or Twitter @jessiet1023 with any comments or questions during the course of the meeting. Better yet, stop me in the lobby and say hello.

Thank you for spending your precious travel dollars to be here with us, and for helping to make the Joint Summits an extraordinary conference once again.

**Jessie D. Tenenbaum, PhD**
Scientific Program Committee Chair
2013 TBI Summit
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Join Sidra Medical and Research Center

Sidra Medical and Research Center is a groundbreaking hospital, research and education institution based in Doha, Qatar, that will focus on the health of women and children regionally and globally.

Technology and Innovation
- Sidra aims to be ranked among the most advanced research hospitals in the world with patient-centered approaches, state-of-the-art medical equipment, and sophisticated laboratories to nurture innovation and clinical advancement.
- End-to-end digital information systems will lead to improved clinical outcomes.

Working Environment
- Physicians will work within high-caliber interprofessional teams and as part of a network of world-leading partners, including Weill Cornell Medical College in Qatar.
- Opportunities for research participation will abound for physicians, with 2.8% of Qatar’s annual GDP allocated for funding and promoting research and development.
- Sidra intends to offer physicians with faculty appointments within Weill Cornell protected time for work associated with the post.
- Design and staffing structure, including a high clinical staff-to-patient ratio, will ensure that physicians have everything they need to deliver patient-focused, transformative care.

Benefits
- Substantial benefits package including highly competitive salary (paid tax free in Qatar) plus performance bonuses, executive level fully furnished accommodation, generous paid vacation time and Continuing Professional Development.
- Centered in the heart of the Arabian Gulf, Qatar is a melting pot of old and new, with an unsurpassed standard of living and a diverse expatriate community providing a home away from home.

For more information, visit careers.sidra.org
## TBI SUMMIT-AT-A-GLANCE

### MONDAY, MARCH 18

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<td>Registration Open</td>
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<tr>
<td>8:30 a.m. – 12:00 p.m.</td>
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<td>T04: Part A – Navigating the Funding Landscape: How to Find, Develop, and Write your First Proposal</td>
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<td>Part B – Translational Science Mock Study Section</td>
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<tr>
<td>10:00 a.m. – 10:30 a.m.</td>
<td>Coffee Break</td>
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<tr>
<td>1:30 p.m. – 3:00 p.m.</td>
<td>Opening Session and Keynote Address: Dr. Debbie Nickerson</td>
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<td>3:30 p.m. – 5:00 p.m.</td>
<td>Scientific Sessions</td>
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<tr>
<td>5:00 p.m. – 6:00 p.m.</td>
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<td>5:30 p.m. – 6:30 p.m.</td>
<td>Ethical, Legal, and Social Issues Working Group Business Meeting</td>
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<tr>
<td>7:00 p.m. – 7:30 p.m.</td>
<td>Genomics Working Group Business Meeting</td>
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<td>Time</td>
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<td>7:00 a.m. – 8:30 a.m.</td>
<td>Birds of a Feather Sessions</td>
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<td>TBI-BOF-01: Big Research Data</td>
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<td>TBI-BOF-02: Ethical Foundations – and Challenges – of Trusted Broker Systems</td>
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<td>8:30 a.m. – 10:00 a.m.</td>
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<td>TBI-05: Databases and Infrastructure</td>
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<td>TBI-LB02: Late Breaking Session - Genomic Medicine</td>
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<td>Coffee Break</td>
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<td>10:30 a.m. – 12:00 p.m.</td>
<td>Scientific Sessions</td>
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<td>TBI-07: In Silico Insitu</td>
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<td>TBI-08: New Approaches to Biomarker Discovery</td>
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<td>TBI-09: Mechanistic Biomarker Discovery</td>
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<td>TBI-10: How Can Bio-ontologies Support Clinical and Translational Science?</td>
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<td>1:30 – 3:00 p.m.</td>
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<td>6:30 p.m. – 8:00 p.m.</td>
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<tr>
<td>6:30 p.m. – 8:30 p.m.</td>
<td>Billiards Meet-up</td>
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### WEDNESDAY, MARCH 20

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<tr>
<td>7:00 a.m. – 8:30 a.m.</td>
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<tr>
<td>7:00 a.m. – 8:15 am</td>
<td>TBI-CRI AMIA Town Hall with Dr. Gilad Kuperman and Dr. Kevin Fickenscher</td>
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<tr>
<td>8:30 a.m. – 10:00 a.m.</td>
<td>Keynote Presentation: Dr. Eric Horvitz</td>
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<td>10:30 a.m. – 12:00 p.m.</td>
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<td>TBI-15: Electronic Health Record Data Mining</td>
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<td>CRI-01: EMR Text Processing/Phenotyping 1</td>
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<td>CRI-02: Integrating Governance of Research Informatics and Healthcare IT across an Enterprise: Experiences from the Trenches</td>
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<tr>
<td>12:15 p.m. – 1:15 p.m.</td>
<td>Lunch &amp; Learn - Velos</td>
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<td>1:30 p.m. – 3:00 p.m.</td>
<td>Scientific Sessions</td>
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<td>TBI-16: TBI in Oncology</td>
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<td>TBI-17: Pharmacogenomics Decision Support</td>
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<td>CRI-03: Silver Medallion Poster Preview</td>
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<td>CRI-04: EMR Text Processing/Phenotyping 2</td>
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<tr>
<td>3:30 p.m. – 5:00 p.m.</td>
<td>Closing Session and TBI Year in Review: Dr. Russ Altman</td>
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<td>5:00 p.m. – 6:00 p.m.</td>
<td>CRI Poster Session 1 and Reception</td>
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<tr>
<td>6:00 p.m. – 7:30 p.m.</td>
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<td>6:00 p.m. – 8:00 p.m.</td>
<td>i2b2 Academic user’s Group 9AUG) Workshop (Affiliate Event)</td>
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<td>6:00 p.m. – 9:00 p.m.</td>
<td>The Ohio State University Red &amp; Grey Reception (Affiliate Event) by invitation</td>
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This year, AMIA offers another stellar Joint Summits on Translational Science, TBI-CRI, March 18 – 22, San Francisco. TBI was the first significant meeting AMIA had created to serve the ever growing population of academicians and researchers interested in translational bioinformatics. Back then, we didn’t know exactly how the role of genomics in personalized medicine would accelerate. Shortly thereafter we added CRI which coincided roughly with the advent of the United States National Institutes of Health calling informatics a compulsory component of their burgeoning CTSA program. We didn’t know that big data and EHR would be central to clinical research informatics in quite the way it is now.

Each year the Joint Summits has been more successful. The meeting AMIA created serves a unique audience and many valued AMIA members and features highly specialized content and top speakers. This year’s program was expertly conceptualized and designed by the Scientific Program Committee, led by Chairs, Jessica D. Tenenbaum, PhD (TBI) and Elmer V. Bernstam, MD, MSE (CRI).

That AMIA can be the professional home for informaticians in translational bioinformatics, clinical research, applied clinical, consumer health, and public health informatics is a remarkably rich accomplishment. While we are far from perfect in serving the unique needs of very different constituencies, our actively engaged members push us to make AMIA and informatics even more influential. What makes AMIA a place for leaders and leadership is our ability to weave together our interconnected, multidisciplinary threads.

AMIA is on a growth trajectory—not by abandoning our traditional strengths in academic research, but by planting roots in that solid foundation and branching outward to embrace the full range of possibility for informaticians across the multidisciplinary spectrum of biomedical informatics.

I welcome your comments, questions and concerns about the future of AMIA. An engaged membership is the lifeblood of the organization! Join us at the Town Hall and speak up or email your thoughts to feedback@amia.org.

Kevin
Kevin M. Fickenscher, MD
President and CEO, AMIA

How Can AMIA Best Support the Goals of Translational Bioinformatics and Clinical Research Informatics?

WEDNESDAY, MARCH 20 7:00 a.m. – 8:15 a.m.

As AMIA grows, the informatics tent grows larger to include the spectrum of domains from translational bioinformatics and clinical research informatics to applied clinical and operational informatics. Members throughout the community have expressed interest in a strategic vision that is rooted in the core research and science interests and also includes expanded outreach and services for applied clinical informaticians.

Join Gilad J. Kuperman, MD, PhD, AMIA Board Chair, and Kevin M. Fickenscher, MD, AMIA President and CEO, for an open Town Hall event with Q&A.

The AMIA leadership is listening and welcomes full attendance at this session. Topics will include work of the Board of Directors, new and continuing initiatives, and an update on AMIA’s strategic directions.
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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.

LEARNING OBJECTIVES
• To present the state-of-the art in biomedical informatics approaches, theories, and methods relevant to clinical and translational sciences
• To present the latest research and development findings on using informatics approaches to improve clinical and translational biomedical research
• To demonstrate frameworks for deploying and assessing clinical research informatics initiatives
• To explore interactions among professionals engaged in clinical and translational science including clinical and translational investigators, computational biologists, genomics researchers, statistical geneticists, clinical informaticians, public health professionals, and those involved with clinical and research IT policy and regulatory issues
• To provide a platform to discuss research-related issues among the national and international clinical and translational research informatics initiatives, such as CTSA, caBIG, CDISC, etc.

For information on how to claim your credits, please go to http://www.amia.org/jointsummits2013/cme
LATE BREAKING SESSIONS

MONDAY, MARCH 18 | 3:30 p.m. – 5:00 p.m. | Room: Cyril Magnin III

**TBI-LB01: Late Breaking Session – Data Integration for Disease Reclassification**

**Network-based Stratification of Tumor Mutations**  
M. Hofree, J. Shen, T. Ideker, University of California San Diego

**Network Models of GWAS Uncover the Topological Centrality of Protein Interactions in Complex Disease Traits**  

**Phenome-wide Analysis of GWAS-discovered Genetic Associations**  
J. Denny, Vanderbilt University

**Integration of Multi-layers of Genomic Data for Cancer Clinical Outcome Prediction**  
D. Kim, H. Shin, Y. Song, J. Kim, Seoul National University

TUESDAY, MARCH 19 | 8:30 a.m. – 10:00 a.m. | Room: Mission

**TBI-LB02: Late Breaking Session – Genomic Medicine**

**Thoughts from SNP-SIG: Future Challenges in the Annotation of Genetic Variations**  
Y. Bromberg, Rutgers University; E. Capriotti, University of Alabama at Birmingham

**Collective Computational Wisdom to Predict Disease-associated SNPs**  
E. Capriotti, University of Alabama at Birmingham; R. Altman, Stanford University; Y. Bromberg, Rutgers University

**CAGI: The Critical Assessment of Genome Interpretation, a Community Experiment to Evaluate Phenotype Prediction**  
S. Brenner, University of California Berkley; S. Repo, EMBL-EMBI; J. Moult, University of Maryland

**Precision Medicine through Finding the True Names of Disease**  
I. Kohane, Boston Children’s Hospital and Harvard Medical School

TUESDAY, MARCH 19 | 1:30 p.m. – 3:00 p.m. | Room: Cyril Magnin I

**TBI-LB03: Late Breaking Session – That’s my Data! Navigating ownership, Sharing, and Translation in the Genomic Era**

K. Edwards, University of Washington; H. Greely, Stanford University; B. Koenig, University of California San Francisco; A. Torrance, University of Kansas

In this era of big data, genomic medicine is enabling personalized approaches to diagnostics and therapeutics in ways never before possible. But with these advances come critical ethical, legal, and social issues around data use. Who owns this data and the new knowledge to which it gives rise? What obligations do researchers have toward participants? What information should researchers be sharing with patients, participants, and society more broadly? Are there novel models of partnership and engagement that can build trust? Where does nature end and intellectual property begin? How are parallel efforts on the consumer market disrupting usual clinical care?

These questions must be addressed by researchers and society alike if we are to harness the power of genomic and large-scale clinical data capabilities in ways that are trustworthy and impactful. In this panel, legal, social, and ethics scholars will speak to issues of data sharing, community engagement, returning results, gene patenting and ownership, and direct-to-consumer movements. Each member of the panel consults actively on large-scale data-driven research projects and is at the front lines of shaping governance processes that will facilitate, rather than hinder, our ability to utilize big data to impact health. In order for the best technology and science in the world to come to bear on human health issues, partners across clinical groups, researchers, policymakers, and society at large must be engaged. This panel discussion will frame emerging issues in the field with researchers and advance further stakeholder engagement.
2013 JOINT SUMMITS
KEYNOTE SPEAKERS

MONDAY, MARCH 18, 2013
1:30 p.m. – 3:00 p.m.  |  Room: Cyril Magnin I/II

DEBBIE NICKERSON, PhD
Professor of Genome Sciences, University of Washington

Debbie Nickerson, PhD, is a Professor in the Department of Genome Sciences and Director of the Northwest Genomics Center at the University of Washington in Seattle, WA, USA. She coordinates the NHGRI U.S. Centers for Mendelian Genomics, and leads the Mendelian Center at the UW. She also leads the NHLBI Exome Sequencing Project, which is a U.S. effort to explore genotype-phenotype associations for cardiovascular and lung diseases and quantitative traits using exome variation. The current focus of her research is on the application of next-generation sequencing in complex human diseases, rare Mendelian disorders and in Pharmacogenomics. She has pioneered the development of new methods and tools that have been widely adopted for the identification and genotyping of human sequence variation, including exome sequencing, and has applied these to uncover the genetic basis of both complex and Mendelian disorders.

TUESDAY, MARCH 19, 2013
3:30 p.m. – 5:00 p.m.  |  Room: Cyril Magnin I/II

STEPHEN FRIEND, MD, PhD
President, Co-Founder and Director of Sage Bionetworks

Dr. Friend is the President of Sage Bionetworks. He is an authority in the field of cancer biology and a leader in efforts to make large scale, data-intensive biology broadly accessible to the entire research community. Dr. Friend has been a senior advisor to the NCI, several biotech companies, a Trustee of the AACR and is a AAAS and Ashoka Fellow as well as an editorial board member of Open Network Biology. Dr. Friend was previously Senior Vice President and Franchise Head for Oncology Research at Merck & Co., Inc. where he led Merck’s Basic Cancer Research efforts.

Prior to joining Merck, Dr. Friend was recruited by Dr. Leland Hartwell to join the Fred Hutchinson Cancer Research Center’s Seattle Project, an advanced institute for drug discovery. While there Drs. Friend and Hartwell developed a method for examining large patterns of genes that led them to co-found Rosetta Inpharmacins in 2001. Dr. Friend has also held faculty positions at Harvard Medical School from 1987 to 1995 and at Massachusetts General Hospital from 1990 to 1995. He received his B.A. in philosophy, his Ph.D. in biochemistry and his M.D. from Indiana University.
**ERIC HORVITZ, MD, PhD**

*Deputy Managing Director at Microsoft Research*

Dr. Horvitz is interested in principles of sensing, learning, and decision making under uncertainty. His interests include computational models of perception, reflection, and action. Beyond theoretical models, he pursues the development and fielding of applications in several realms, including time-critical decisions, information retrieval, healthcare, urban infrastructure, sustainability, and development—with goals of understanding how computational models perform amidst real-world complexities, and of deploying systems that deliver value to people and society.

Related interests include machine learning and decision making for crowdsourcing and human computation, information triage and alerting that takes human attention into consideration, spanning work on notification systems, surprise modeling, multitasking, and psychological studies of interruption and recovery. Other interests include principles of mixed-initiative interaction that can support fluid, efficient collaborations between people and computing systems, methods for guiding computer actions in accordance with the preferences of people, search and information retrieval, and collaboration.

Theoretical research interests include offline and real-time optimization of the expected value of computational systems under limited and varying resources. Areas of concentration in this realm include flexible or anytime computation, ideal metareasoning for guiding computation, compilation for reducing real-time deliberation, ongoing, continual computation, and the construction of bounded-optimal reasoning systems—systems that maximize the expected utility of the people they serve, given the expected costs of reasoning, the problems encountered over time, and assertions about a system's constitution. Research in this arena includes tackling hard reasoning problems with learning and decision making methods.

**RUSS B. ALTMAN, MD, PhD**

*Kenneth Fong Professor of Bioengineering and Genetics, Stanford University*

Dr. Altman is also a Professor of Medicine & (by courtesy) Computer Science and Director, Biomedical Informatics Training Program Stanford University. He focuses on the creation and application of computational tools to solve problems in biology and medicine. Current application projects include the study of structure-function relationships in macromolecular structure, understanding the structure and folding of RNA molecules, and analyzing the relationship of genotype and phenotype, particularly with respect to the response to drugs. Techniques used include knowledge representation, database design, machine learning, natural language processing, physics-based simulation and graph-based modeling/analysis.
AMIA members with 20+ years of membership earn a Silver Medallion pin for loyalty and dedication to the AMIA mission.

**MONDAY MARCH 18**
3:30 p.m. – 5:00 p.m.

Room: Mission

Silver Medallion TBI Poster Framing is new this year and 58 posters are included in the TBI Poster Session. So many posters, so little time! This novel session will feature a selection of top posters as presented, contextualized, and discussed by some of the most distinguished researchers and AMIA members in the field of Translational Bioinformatics.

**ATUL BUTTE**, MD, PhD
*Stanford University School of Medicine*

**BRIAN ATHEY**, PhD
*University of Michigan, Computational Medicine and Bioinformatics*

**Isaac Kohane**, PhD, MD
*Harvard Medical School*

**5:00 p.m. – 6:00 p.m.**

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

(See page 28 YELLOW for list of TBI posters, listed alpha order by first author.)

(See page 14 GREEN for information on Silver Medallion CRI Poster Framing.)
**TUESDAY, MARCH 19  TBI**

12:15 p.m. – 1:15 p.m.  |  Room: Cyril Magnin III

**Translational Research Center: A Data Management Platform to Support Value Based, Personalized Medicine**

This presentation will focus on the new analytics solutions that will be required to provide a scalable, secure platform for personalized medicine that accelerates biomarker discovery, validation and ultimately decision making at the point of care. This session will also discuss the challenges associated with integrating cross platform ‘omics’ data in a manner that scales to thousands of whole genome sequences whilst integrating with longitudinal clinical data from EMRs, case reports, registries and other “real world” data sources to provide an integrated view across genotype and phenotype. Specifically, Oracle will demonstrate its Translational Research Center (TRC) platform that is being implemented by many of the world’s leading academic medical centers, integrated delivery networks and cancer centers to answer the “hard questions” in healthcare – what works, for whom, why, in what context and at what cost?

**WEDNESDAY, MARCH 20  TBI-CRI Bridge Day**

12:15 p.m. – 1:15 p.m.  |  Room: Cyril Magnin III

**Clinical Data Management for Adverse Events:** tracking and reporting of events based on Common Toxicity Criteria and Graft-Versus-Host Disease grading standards—using mobile solutions from Velos.

This session will demonstrate Velos’ point of care solution for the management of AEs, aGVHDs, and cGVHDs using Common Toxicity Criteria for Adverse Events (CTCAE) and the NIH Criteria. Velos’ search and report engines that enable users with limited IT knowledge to quickly generate custom reports on data stored in Velos eResearch. Planned use in a study will be demonstrated.

Velos’ trusted Internet-based platform provides research teams with secure and compliant ways of managing clinical research and specialized medical records. With deep investments in R&D, we strive to continually improve our offerings, in order to help our customers reduce costs, increase efficiency and ultimately provide better patient care. Contact: Alex Pike, Business Analyst, apike@velos.com

**THURSDAY MARCH 21  CRI**

12:15 p.m. – 1:15 p.m.  |  Room: Cyril Magnin III

**Michael Kahn, MD, PhD, Associate Professor, Department of Pediatrics, University of Colorado, and Director of Clinical Informatics in the Department of Quality & Patient Safety at The Children’s Hospital, Denver.**

Dr. Kahn will be speaking on his choice of OMOP, the Observational Medical Outcomes Partnership, for the data model and tools used to implement the open source ROSITA server for the SAFTINet project, and the role that Recombinant by Deloitte has played in implementing ROSITA.

**Lisa M. Schilling, MD, MSPH, Associate Professor of Medicine, Division of General Internal Medicine, University of Colorado Health Sciences Center; Director, Evidence-based Medicine and Medical Informatics Curriculum, School of Medicine; and Director, Scalable Architecture for Federated Therapeutic Inquiries Network (SAFTINet).**

Dr. Schilling is the Principle Investigator, SAFTINet, the AHRQ funded effort that is using the OMOP software.
Birds-of-a Feather sessions are informal sessions for professionals to mingle and share experiences.

*(See page 16 GREEN for information about CRI Birds-of-a-Feather Sessions.)*

**TUESDAY, MARCH 19**  
7:00 a.m. – 8:30 a.m.

**TBI-BOF-01: Big Research Data**  
*N. Anderson, University of Washington*

Characteristics of biomolecular data differentiate it from other classes of data commonly found in EHRs, and challenges of management and integration into healthcare settings reflect increased use of often proprietary instrumentation. Extending these data to translational research environments faces challenges of storage, transport and provenance issues, and will require adoption of scientific computing models found both in industry and beyond health sciences.

**TBI-BOF-02: Ethical Foundations – and Challenges – of Trusted Broker Systems**  
*K. Goodman, R. Fiore, University of Miami*

The evolution of trustee models for coordination and management of privacy and consent issues has yet to produce a widely agreed-upon governance structure. Standards are sorely needed to address questions of secondary use, opt in/opt out and other challenges of digital translational bioscience.
MEETINGS/SOCIAL

JILLIAN’S BILLIARDS CLUB
TUESDAY, MARCH 19
Time: 6:30 p.m. – 8:30 p.m.

Jillian’s Billiards Club is warm and welcoming. The Billiards rooms offer a relaxed lounge atmosphere, with plush chairs and sofas, the vibe is easy going, laid back, and most of all fun! Guest tickets may be purchased for $50 at the Joint Summits registration desk.

Location: 175 4th Street, San Francisco. Transportation on your own. Located near the Parc 55 Hotel.

Tickets: Free for Joint Summits registrants. Includes one complimentary drink ticket and heavy hors d’oeuvres.

Socialize and enjoy an evening out with colleagues. Fun, food and FREE entrance for Joint Summits registrants!

WEDNESDAY, MARCH 20
6:00 p.m. – 9:30 p.m.

The Ohio State University Red & Grey Reception (Affiliate Event) by invitation

Room: Cyril Magnin III

MEETINGS

MONDAY, MARCH 18
5:30 p.m. – 6:30 p.m.

Ethical, Legal, and Social Issues Working Group Business Meeting

Room: Cyril Magnin I

TUESDAY, MARCH 19
6:30 p.m. – 8:00 p.m.

tranSMART Community Meeting (Affiliate Event)

Want to know more about tranSMART? Come learn about the Open Source and Community-Driven Informatics and Data Sharing Platform for Clinical and Translational Research. Updates on major projects and the tranSMART roadmap will be presented. Connect with newbie and veteran members of the tranSMART community.

Room: Mission

6:00 p.m. – 7:30 p.m.

Natural Language Processing Working Group Business Meeting

Room: Balboa

6:00 p.m. – 8:00 p.m.

i2b2 Academic Users’ Group (AUG) Workshop (Affiliate Event)

Room: Mission
### DAILY SCHEDULE  
**MONDAY, MARCH 18**

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<tr>
<th>Time</th>
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<th>Room</th>
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<tr>
<td>7:00 a.m. – 8:30 a.m.</td>
<td>Coffee and Pastries</td>
<td>Cyril Magnin Foyer</td>
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<tr>
<td>7:30 a.m. – 5:30 p.m.</td>
<td>Registration Open</td>
<td>Cyril Magnin Foyer</td>
</tr>
<tr>
<td>8:30 a.m. – 12:00 p.m.</td>
<td>Tutorials</td>
<td>Cyril Magnin Foyer</td>
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</table>

Tutorials incur a fee in addition to the applicable registration fee. CME eligibility for the Half-day sessions = 3 CME.

Member fee: $95; Non-member fee: $150. Visit registration to inquire about availability.

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**T01: Introduction to Translational Bioinformatics**  
*Room: Mission I*

Maricel G. Kann, University of Maryland, Baltimore County; Yves A. Lussier, The University of Illinois in Chicago

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: Using ontologies for Data-driven Medicine**  
*Room: Mission II/III*

Nigam H. Shah, Stanford Center for Biomedical Informatics Research; Rave Harpaz, Stanford University

Data-driven medicine envisions the discovery of new treatment options based on the multi-model molecular measurements on patients and learning from the trends hidden among the diagnoses, prescriptions, and discharge summaries of millions of patient encounters logged by clinical practitioners. Biomedical ontologies are widely used across the whole spectrum of this research enterprise. The National Center for Biomedical Ontology (NCBO) enables users to access biomedical terminologies, create lexicons, index the contents of online data sets with controlled terms (data annotation), and to recommend particular ontologies appropriate for specific data-annotation tasks. This tutorial will demonstrate the use of NCBO technology in data mining methods that transform unstructured patient notes taken by doctors, nurses and other clinicians into a de-identified, temporally ordered, patient-feature matrix using standardized medical terminologies. We will review how to use the resulting high-throughput data for earning practice-based evidence, for conducting drug safety studies, and for building predictive models.
**T03: Research Informatics: From Lab to Laptop**  
*Philip R.O. Payne, Peter J. Embi, The Ohio State University*

A common substrate underlying the conduct of trans-disciplinary research is the use of biomedical informatics theories and methods, as well as a variety of computational technologies. Common information needs that are targeted in such contexts include supporting team collaboration, project planning, data management, knowledge generation, and results dissemination. In this tutorial, we will provide researchers, decision makers, and technical staff with an overview of the core definitions, theories, and best practices that collectively contribute to the successful application of biomedical informatics in order to facilitate and enable biomedical research.

**T04: Part A – Navigating the Funding Landscape: How to Find, Develop, and Write your First Proposal**  
*Room: Davidson*

*Russ Altman, Stanford University; Sean Mooney, Buck Institute for Research on Aging*

Your departmental chair has asked you to develop your first funding proposal. In this presentation, the process of finding and writing a topical and fundable proposal will be described. In the first talk, Prof. Altman will describe the keys to writing a proposal narrative that will be seen as successful by the reviewers. The dos and don’ts of proposal writing will be outlined and tips for submitting competitive language of interest to the reviewers will be highlighted.

In the second presentation, Prof. Mooney will describe navigating the funding landscape. The process of finding NSF and NIH opportunities, deciding on a research topic and identifying appropriate reviewers will be noted. The basics of proposal preparation will be illustrated through example, including conversations you will have with your university administrators and NIH program staff. The resources to find funding opportunities will be distributed.

**T04: Part B – Translational Science Mock Study Section**  
*Room: Davidson*

*Shannon McWeeney, Oregon Health & Science University; Nick Anderson, University of Washington*

What just happened to my grant? How can I benefit from the grant peer review process? In this workshop, attendees will have the opportunity to learn about the proposal review process, including the typical dynamics of study section deliberations. The NIH study section review process will be demonstrated, and the workshop will present examples of mistakes applicants commonly make.

**10:00 a.m. – 10:30 a.m.  Coffee Break**  
*Room: Cyril Magnin Foyer*

*(Lunch on your own)*

**1:30 p.m. – 3:00 p.m.  TBI Opening Session and Keynote Presentation**  
*Room: Cyril Magnin I/II*

**DEBBIE NICKERSON, PhD**  
*Professor of Genome Sciences, University of Washington*

*(See page 12 for description.)*
3:00 p.m. – 3:30 p.m.  Coffee Break  
Room: Cyril Magnin Foyer

3:30 p.m. – 5:00 p.m.  Scientific Sessions

**TBI-01: Panel – Developing, Implementing, and Sharing Pharmacogenomic Clinical Decision Support**  
Room: Cyril Magnin I

J. Hoffman, St. Jude Children’s Research Hospital; J. Starren, Northwestern University Feinberg School of Medicine; J. Peterson, Vanderbilt University Medical Center; M. Hoffman, Cerner Corporation; R. Freimuth, Mayo Clinic

As pharmacogenomic knowledge is translated into clinical practice, clinical decision support (CDS) in the electronic health record (EHR) will play a crucial role in maximizing the use of pharmacogenomic data over a patient’s lifetime. CDS must be designed to enable clinicians to understand and act upon patient-specific pharmacogenomic data. To accelerate the adoption of pharmacogenomic CDS, the significant effort and lessons learned at innovator sites must be organized and shared among care settings and EHR vendors. While best practices are emerging to represent and share CDS across sites, unique considerations exist for sharing CDS related to genomic data. Panelists from early adopter institutions will compare and contrast their experiences developing and implementing CDS for pharmacogenomics. Perspectives on the integration of pharmacogenomic data into commercial EHRs will be given by a major EHR vendor, and matters specific to knowledge management and sharing CDS for pharmacogenomics in human and machine readable forms will be discussed. Key implementation issues that will be addressed by the panelists include the role of CDS in ordering pharmacogenomics panel tests, the integration of genomic and other clinical data, and the long-term management of genomic data in the EHR.

**TBI-02: Papers/Podium Presentations - Genotypes and Phenotypes**  
Room: Cyril Magnin II

Session Chair: Vanathi Gopalakrishnan

- **Anonymous Record Linkage between EPR and CDW-H: Toward Development of a Federated Genotype-phenotype System**
  D. Pu, J. Mostafa, University of North Carolina at Chapel Hill; S. Garantziotis, National Institute of Environmental Health Sciences

- **Autworks: A Cross-disease Analysis Application for Autism and Related Disorders**
  D. Wall, Harvard University

- **Genetic Networks of Complex Disorders: From a Novel Search Engine for PubMed Article Database**
  J. Jung, D. Wall, Harvard Medical School

- **Standardizing Phenotype Variables in the Database of Genotypes and Phenotypes (dbGaP) based on Information Models**
  K. Lin, A. Hsieh, S. Farzaneh, S. Doan, H. Kim, University of California, San Diego

Tweet your Session  
#TBICRI13
**TBI-03: TBI Silver Medallion Poster Framing**  
*PREVIEW*  
Room: Mission

A. Butte, Stanford University; B. Athey, University of Michigan; I. Kohane, Harvard Medical School

New this year, 58 posters are included in the TBI Poster Session. So many posters, so little time! This novel session will feature a selection of top posters as presented, contextualized, and discussed by some of the most distinguished researchers and AMIA members in the field of Translational Bioinformatics.

*(See page 28 YELLOW for list of posters, listed alpha order by first author.)*

**TBI-LB01: Late Breaking Session - Data Integration for Disease Reclassification**  
Room: Cyril Magnin III

Session Chair: Maricel Kahn

**Network-based Stratification of Tumor Mutations**  
M. Hofree, J. Shen, T. Ideker, University of California San Diego

**Network Models of GWAS Uncover the Topological Centrality of Protein Interactions in Complex Disease Traits**  

**Precision Medicine through Finding the True Names of Disease**  
I. Kohane, Boston Children’s Hospital and Harvard Medical School

**Integration of Multi-layers of Genomic Data for Cancer Clinical Outcome Prediction**  
D. Kim, H. Shin, Y. Song, J. Kim, Seoul National University

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**5:00 p.m. – 6:00 p.m. Reception**  
Room: Cyril Magnin Foyer

**5:30 p.m. – 6:30 p.m. Business Meeting**  
Genomics Working Group Business Meeting  
Room: Cyril Magnin II

**6:00 p.m. – 7:00 p.m. Business Meeting**  
Ethical Legal and Social Working Group Business meeting  
Room: Cyril Magnin I
7:00 a.m. – 8:30 a.m.  Coffee and Pastries

7:00 a.m. – 8:30 a.m.  Birds-of-a-Feather Session

**TBI-BOF-01: Big Research Data**

*Room: Cyril Magnin I*

N. Anderson, *University of Washington*

Characteristics of biomolecular data differentiate it from other classes of data commonly found in EHRs, and challenges of management and integration into healthcare settings reflect increased use of often proprietary instrumentation. Extending these data to translational research environments faces challenges of storage, transport and provenance issues, and will require adoption of scientific computing models found both in industry and beyond health sciences.

**TBI-BOF-02: Ethical Foundations – and Challenges – of Trusted Broker Systems**

*Room: Cyril Magnin II*

K. Goodman, R. Fiore, *University of Miami*

The evolution of trustee models for coordination and management of privacy and consent issues has yet to produce a widely agreed-upon governance structure. Standards are sorely needed to address questions of secondary use, opt in/opt out and other challenges of digital translational bioscience.

8:30 a.m.-10:00 a.m.  Scientific Sessions

**TBI-04: Panel - tranSMART: An Open Source and Community-driven Informatics and Data Sharing Platform for Clinical and Translational Research**

*Room: Cyril Magnin I*

B. Athey, *University of Michigan*; M. Braxenthaler, Pistoia Alliance; M. Haas, *One Mind for Research*; Y. Guo, Imperial College London

tranSMART is an emerging global open source public private partnership community developing a comprehensive informatics-based analysis and data-sharing cloud platform for clinical and translational research. The tranSMART consortium includes pharmaceutical and other companies, not-for-profits, academic entities, patient advocacy groups, and government stakeholders. The tranSMART value proposition relies on the concept that the global community of users, developers, and stakeholders are the best source of innovation for applications and for useful data. Continued development and use of the tranSMART platform will create a means to enable “pre-competitive” data sharing broadly, saving money and, potentially accelerating research translation to cures. Significant transformative effects of tranSMART includes 1) allowing for all its user community to benefit from experts globally, 2) capturing the best of innovation in analytic tools, 3) a growing ‘big data’ resource, 4) convergent standards, and 5) new informatics-enabled translational science in the pharma, academic, and not-for-profit sectors.

**TBI-05: Papers/Podium Presentations - Databases and Infrastructure**

*Room: Cyril Magnin II*

Session Chair: Subha Madhaven

Harvest: A Web-based Biomedical Data Discovery and Reporting Application Development Platform

Biotrust: A Comprehensive System for Acquiring and Distributing Biospecimens
N. Anderson, S. Ponkos, R. Black, J. Prosser, University of Washington; B. Stein, Fred Hutchinson Cancer Research Center; P. Tarczy-Hornoch, University of Washington; P. Porter, Fred Hutchinson Cancer Research Center

The Stem Cell Commons: An Exemplar for Data Integration in the Biomedical Domain Driven by the ISA Framework
S. Ho Sui, Harvard School of Public Health; E. Merrill, Massachusetts General Hospital; N. Gehlenborg, P. Haseley, Harvard Medical School; I. Sytczay, Harvard School of Public Health; R. Park, Harvard Medical School; P. Rocca-Serra, University of Oxford; S. Corlosquet, Massachusetts General Hospital; A. Gonzalez-Beltran, E. Maguire, University of Oxford; O. Hofmann, Harvard School of Public Health; P. Park, Harvard Medical School; S. Das, Massachusetts General Hospital; S. Sansone, University of Oxford; W. Hide, Harvard School of Public Health

Development of an Open-source, Flexible Framework for Complex Inter-institutional Disparate Data Sharing and Collaboration
S. Volchenboum, C. Kirby, University of Chicago; P. Ambros, Children’s Cancer Research Institute; D. Billiter, The Research Institute at Nationwide Children’s Hospital; W. London, Dana-Farber Cancer Institute; E. Mendonca, University of Wisconsin-Madison; T. Monclair, Rikshospitalet University Hospital; A. Pearson, Institute of Cancer Research and Royal Marsden Hospital; S. Cohn, University of Chicago

TBI-06: Papers/Podium Presentations - Analytic Pipelines
Room: Cyril Magnin III
Session Chair: Sean Mooney

SysBioCube: A Data Warehouse and Integrative Data Analysis Platform Facilitating Systems Biology Studies of Disorders of Military Relevance
S. Chowbina, SAIC

Clinical Genomicist Workstation

Galaxy High Throughput Genotyping Pipeline for GeneTitan
N. Bahroos, M. Chukhman, UIC; X. Dong, Center for Clinical and Translational Science; O. Karpenko, Z. Arbieva, T. Jackson, W. Hendrickson, UIC

Genome and Proteome Annotation using Automatically Recognized Concepts and Functional Networks
A. Bivol, T. Wittkop, D. Davis, S. Mooney, Buck Institute for Research on Aging

TBI-LB02: Late Breaking Session - Genomic Medicine
Room: Mission
Session Chair: Maricel Kahn

Thoughts from SNP-SIG: Future Challenges in the Annotation of Genetic Variations
Y. Bromberg, Rutgers University; E. Capriotti, University of Alabama at Birmingham

Collective Computational Wisdom to Predict Disease-associated SNPs
E. Capriotti, University of Alabama at Birmingham; R. Altman, Stanford University; Y. Bromberg, Rutgers University

CAGI: The Critical Assessment of Genome Interpretation, a Community Experiment to Evaluate Phenotype Prediction
S. Brenner, University of California Berkley; S. Repo, EMBL-EMBL; J. Moult, University of Maryland

Phenome-wide Analysis of GWAS-discovered Genetic Associations
J. Denny, Vanderbilt University
### TBI SCIENTIFIC SESSIONS | CONTINUED

**10:00 a.m. – 10:30 a.m.**  
**Coffee Break**  
*Room: Cyril Magnin Foyer*

**10:30 a.m. – 12:00 p.m.**  
**Scientific Sessions**  

**TBI-07: In Silico Insitu**  
*Session Chair: Zhan Ye*  

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<td>Drug Repositioning from the Combined Evaluation of Phenotypic and Target-based Screening</td>
<td>S. Swamidass, Washington University; P. Agarwal, GlaxoSmithKline</td>
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<tr>
<td>PharmGKB Drug Data Normalization with NDF-RT</td>
<td>Q. Zhu, R. Freimuth, J. Pathak, C. Chute, Mayo Clinic</td>
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<td>Development of an Ensemble Resource Linking MEDications to their Indications (MEDI)</td>
<td>W. Wei, R. Cronin, H. Xu, T. Lasko, Vanderbilt University; L. Bastarache, J. Denny, Vanderbilt School of Medicine</td>
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**TBI-08: Papers/Podium Presentations – Diverse Approaches to Biomarker Discovery**  
*Session Chair: Barbara Engelhardt*  

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<tr>
<td>Mining Patterns Among Adverse Events in Clinical Trials - An Exploratory Study</td>
<td>Z. Luo, G. Zhang, R. Xu, Case Western University</td>
</tr>
<tr>
<td>A Probabilistic Model of Functional Brain Connectivity Network for Discovering Novel Biomarkers</td>
<td>J. Bian, University of Arkansas for Medical Sciences; M. Xie, University of Arkansas at Little Rock; J. Cisler, University of Arkansas for Medical Sciences</td>
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<tr>
<td>Order-Disorder Interface Characterization Reveals Critical Factors for Disease and Drug Targets</td>
<td>J. Kallenbach, Harvard Medical School; W. Hsu, Indiana University School of Medicine; G. Alterovitz, Harvard Medical School</td>
</tr>
<tr>
<td>New Models of Clinical Discovery &amp; Science: Progress and Lessons Learned the Front Lines of the Department of Veterans’ Affairs Transformational Initiatives</td>
<td>L. D’Avolio, L. Fiore, VA Boston Healthcare System</td>
</tr>
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**TBI-09: Papers/Podium Presentations – Mechanistic Biomarker Discovery**  
*Session Chair: Josh Stuart*  

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<tr>
<th>Title</th>
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<tr>
<td>Outlier Detection through Bipartite Visual Analytics</td>
<td>J. Drake, B. Dang, University of Texas Medical Branch; S. Visweswaran, University of Pittsburgh; S. Bhavnani, University of Texas Medical Branch</td>
</tr>
<tr>
<td>Using Systems Biology-based Analysis Approaches to Identify Mechanistically Significant Adverse Drug Reactions: Pulmonary Complications from Combined Use of Anti-TNFα Agents and Corticosteroids</td>
<td>M. Sarangdhar, A. Kushwaha, J. Dahliquist, A. Jegga, B. Aronow, Cincinnati Children’s Hospital Medical Center</td>
</tr>
<tr>
<td>Network-based Target Ranking for Polypharmacological Therapies</td>
<td>F. Vitali, F. Mulas, University of Pavia; P. Marini, Demetra Pharmaceutical; R. Bellazzi, Università di Pavia</td>
</tr>
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</table>
How Cytokines Co-occur across Rickettsioses Patients: From Bipartite Visual Analytics to Mechanistic Inferences of a Cytokine Storm
S. Bhavnani, J. Drake, University of Texas Medical Branch; G. Bellala, Hewlett Packard Laboratories; B. Dang, B. Peng, University of Texas Medical Branch; J. Oteo, P. Santibanez, Hospital San Pedro; S. Visweswaran, University of Pittsburgh; J. Olano, University of Texas Medical Branch

TBI-10: Panel - How can Bio-ontologies Support Clinical and Translational Science?
Room: Cyril Magnin I
W. Hogan, UAMS; N. Shah, Stanford University; W. Kibbe, Northwestern University; M. Haendel, Oregon Health & Sciences University; M. Brochhausen, University of Arkansas for Medical Sciences

In this panel we will present state-of-the-art applications of ontology to translational science, and derive from that experience what we can learn regarding principles for the future development and use of ontologies to promote translational science. Towards that end, panelists will describe existing benefits of cutting-edge, ontology-driven computing for translational science as well as the pressing issues that require resolution, and how these early results inform the principles we might follow moving forward. We anticipate that a diverse audience will benefit from the discussion: informaticists and researchers will learn the value that ontologies can provide to their research as well as how to choose and integrate them into their work; system developers will learn how to leverage ontologies in support of scientific research; and ontology developers will have a chance to hear and participate in the discussion of proposals for principles for ontology development and use. The intended audience for this panel is therefore biomedical researchers, informaticists, ontology researchers, ontology users, health sciences and informatics students, and health information and knowledge management professionals, especially from CTSA sites.

12:15 p.m. – 1:15 p.m. Lunch & Learn
Room: Cyril Magnin III
Oracle Health Sciences
Translational Research Center: A Data Management Platform to Support Value Based, Personalized Medicine
(See page 15 YELLOW for description.)

1:30 p.m. – 3:00 p.m. Scientific Sessions
Room: Cyril Magnin I
TBI-11: Papers/Podium Presentations – Research on Research
Session Chair: Harold “Skip” Garner

clearScience: Infrastructure for Communicating Data-Intensive Science
E. Huang, B. Bot, D. Burdick, M. Kellen, Sage Bionetworks

Educating Translational Researchers in Research Informatics Principles and Methods: An Evaluation of a Model Online Course and Plans for its Dissemination
R. Friedman, Boston Medical Center; V. Kudesia, Boston University Medical Center; P. Sebastiani, Boston University School of Public Health; S. Monti, Boston University School of Medicine; D. Misquitta, Boston University Medical Center; K. Peterson, J. Whinfield, R. Stoeckle, Education Development Centers, Inc.

Adoption of Research Networking Systems in the Clinical and Translational Science Award (CTSA) Consortium
L. Johnson, University of Minnesota; S. Stallings, Vanderbilt University; D. Eichmann, University of Iowa; J. Obeid, Medical University of SC

Genomes in the Cloud: Balancing Privacy Rights and the Public Good
L. Ohno-Machado, C. Farcaș, J. Kim, S. Wang, X. Jiang, University of California San Diego
Learning Drug-drug Interactions from the Unstructured Text of Electronic Health Records

Extending the "Web of Drug Identity" with Knowledge Extracted from United States Product Labels
O. Hassanzadeh, IBM Research; Q. Zhu, R. Freimuth, Mayo Clinic; R. Boyce, University of Pittsburgh

Pharmacovigilance using Clinical Text

Categorizing Medications from Unstructured Clinical Notes
F. Farooq, Siemens Medical Solutions USA, Inc.; S. Yu, B. Krishnapuram, V. Anand, Siemens Healthcare

Robust Prediction-based Analysis for Genome-wide Association and Expression Studies
S. Koppula, A. Zollanvari, G. Alterovitz, Massachusetts Institute of Technology

Systematic Identification of Interaction Effects between Validated Genome- and Environment-wide Associations on Type 2 Diabetes Mellitus
C. Patel, R. Chen, K. Kodama, J. Ioannidis, A. Butte, Stanford University

Analysis of Sequence-based Copy Number Variation Detection Tools for Cancer Studies
S. Nabavi, P. Tonellato, Z. Cai, Harvard Medical School

An Empirical Workflow for Genome-wide Single Nucleotide Polymorphism-based Predictive Modeling
C. Floudas, J. Balasubramanian, M. Romkes, V. Gopalakrishnan, University of Pittsburgh

Navigating Ownership, Sharing, and Translation in the Genomic Era
K. Edwards, University of Washington; H. Greely, Stanford University; B. Koenig, University of California San Francisco; A. Torrance, University of Kansas

In this era of big data, genomic medicine is enabling personalized approaches to diagnostics and therapeutics in ways never before possible. But with these advances come critical ethical, legal, and social issues around data use. Who owns this data and the new knowledge to which it gives rise? What obligations do researchers have toward participants? What information should researchers be sharing with patients, participants, and society more broadly? Are there novel models of partnership and engagement that can build trust? Where does nature end and intellectual property begin? How are parallel efforts on the consumer market disrupting usual clinical care? These questions must be addressed by researchers and society alike if we are to harness the power of genomic and large-scale clinical data capabilities in ways that are trustworthy and impactful. In this panel, legal, social, and ethics scholars will speak to issues of data sharing, community engagement, returning results, gene patenting and ownership, and direct-to-consumer movements. Each member of the panel consults actively on large-scale data-driven research projects and is at the front lines of shaping governance processes that will facilitate, rather than hinder, our ability to utilize big data to impact health. In order for the best technology and science in the world to come to bear on human health issues, partners across clinical groups, researchers, policymakers, and society at large must be engaged. This panel discussion will frame emerging issues in the field with researchers and advance further stakeholder engagement.
10:00 a.m. – 10:30 a.m. Coffee Break
Room: Cyril Magnin Foyer

3:30 p.m. – 5:00 p.m. Keynote Presentation
Room: Cyril Magnin I/II

STEPHEN FRIEND, MD, PhD
President, Co-Founder and Director of Sage Bionetworks

(See page 12 YELLOW for description.)

5:00 p.m. – 6:00 p.m. TBI Poster Session and Reception
Room: Cyril Magnin Foyer

(Purchase tickets at Registration)

(See page 28 YELLOW for list of posters, listed alpha order by first author.)

6:30 p.m. – 8:00 p.m. Affiliate Event
Room: Mission

tranSMART Community Meeting
Want to know more about tranSMART? Come learn about the Open Source and Community-Driven Informatics and Data Sharing Platform for Clinical and Translational Research. Updates on major projects and the tranSMART roadmap will be presented. Connect with newbie and veteran members of the tranSMART community.

6:30 p.m. – 8:00 p.m. Billiards Meet-up
Jillian’s Billiards Club
175 Fourth Street
San Francisco

Socialize and enjoy an evening out with colleagues. Fun, food and FREE entrance for Joint Summits registrants! Includes one complimentary drink ticket and heavy hors d’oeuvres. Additional guest tickets: $50. (Purchase tickets at Registration)

Jillian’s Billiards Club is warm and welcoming. The Billiards rooms offer a relaxed lounge atmosphere, with plush chairs and sofas, the vibe is easy going, laid back, and most of all FUN! Transportation on your own. Near to Parc 55 Hotel.
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| 5:00 p.m. – 6:00 p.m. | Room: Cyril Magnin Foyer | Process Automation for Efficient Translational Research on Endometrioid Ovarian Carcinoma  
M. Ahuja, T. Bair, M. Goodheart, B. Knosp, University of Iowa  
Board 1  
Characterizing Protein Disorder-order Interactions in Disease  
G. Anand, Harvard Medical School; W. Hsu, Indiana University School of Medicine; G. Alterovitz, Harvard-MIT Division of Health Science  
Board 2  
Data Mining of the Human Gut Microbiota  
T. Altman, Stanford University  
Board 3  
Best Practices in Biomedical Data Extraction, Transformation and Load  
D. Aronzon, M. Palchuk, Recombinant Data Corp.  
Board 4  
Next Generation Cyberinfrastructures for Next Generation Sequencing and Genome Science  
W. Barnett, R. LeDuc, Indiana University  
Board 5  
The MURDOCK Study: A Rich Data Resource for Biomarker Discovery  
Board 6  
How Complimentary Bipartite Visual Analytical Representations Reveal Patterns in Cytokine Expression: A Case Study in Rickettsioses  
S. Bhavnani, B. Dang, J. Drake, University of Texas Medical Branch; G. Bellala, Hewlett Packard Laboratories; J. Olano, University of Texas Medical Branch  
Board 7  
Clustering of Somatic Mutations to Characterize Cancer Heterogeneity with Whole Genome Sequencing  
J. Becq, A. Alexa, R. Grocock, K. Cheetham, Z. Kingsbury, S. Humphray, D. McBride, Illumina UK; A. Timbs, Oxford Radcliffe NHS Trust; A. Schuh, Oxford Cancer and Haematology Centre; M. Ross, Bentley, Illumina UK  
Board 8  
From Genes to Pathways and Back Again: A Visual Analytical Approach  
B. Dang, J. Drake, University of Texas Medical Branch; G. Bellala, Hewlett Packard Laboratories; S. Bhavnani, University of Texas Medical Branch  
Board 9  
Comprehension of Multiple Molecular Pathways using 3D Networks  
J. Drake, B. Dang, University of Texas Medical Branch; S. Visweswaran, University of Pittsburgh; J. Olano, S. Bhavnani, University of Texas Medical Branch  
Board 10  
Big Data in the Research Domain: The Power of a Big Data Appliance to Manage Genomic and EHR Data  
J. Cowan, M. Basford, Vanderbilt University; R. Carroll, Vanderbilt University Medical Center; P. Harris, B. Malin, T. Lasko, Vanderbilt University; J. Denny, Vanderbilt School of Medicine  
Board 11  
Measuring “Unlikely” Gene Similarity Using Weighted Random Sampling  
D. Davis, S. Mooney, T. Wittkop, Buck Institute for Research on Aging  
Board 12 |
Extracting Interacting Orthologs from Literature  
E. Doughty, R. Mishra, M. Kann, University of Maryland, Baltimore County  
Board 13

Quantitative Characterization of Transcriptional Responses to Engineered Nanoparticles of Different Physico-chemical Parameters  
J. Drocco, A. Nagy, R. Iyer, Los Alamos National Laboratory  
Board 14

Phylomedicine of Adverse Drug Events  
J. Dudley, Mount Sinai School of Medicine  
Board 15

Predicting Antigenic Similarity from Sequence for Influenza Vaccine Strain Selection  
J. Espino, S. Visweswaran, M. Wagner, University of Pittsburgh  
Board 16

Targeting the Root of Cancer: A Data-driven Approach for Identification of Cancer Stem Cell Biomarkers  
H. Fan-Minogue, Stanford University; J. Wheeler, Google; A. Butte, Stanford University  
Board 17

Pharmacogenomic Drug Dosing Guidelines: A Model to Support Adoption  
R. Freimuth, C. Chute, Mayo Clinic  
Board 18

Bioinformatics Needs Assessment and Support for Clinical and Translational Science Research  
Board 19

Inferring Medical Diagnoses from Patient Similarities  
A. Gottlieb, Stanford University; G. Stein, Tel Aviv University & Bellinson Hospital/Rabin Medical Center; R. Altman, Stanford University; R. Shahran, Tel Aviv University  
Board 20

Phenotype-genotype Integrator (PheGenI) Updates: Synthesizing Genome-wide Association Study (GWAS) Data with Existing Genomic Resources  
L. Hindorff, NHGRI, NIH; D. Hoffman, NCBI, NIH; H. Junkins, NHGRI, NIH; M. Kimura, D. Maglott, L. Phan, S. Sherry, M. Feolo, NCBI, NIH; E. Ramos, NHGRI, NIH  
Board 21

VIVO: Connect, Share, Discover  
K. Holmes, Washington University School of Medicine; M. Conlon, University of Florida; J. Corson-Rikert, Cornell University  
Board 22

Gene Ontology Annotation Prediction for GWAS Genes using Protein-Protein Interactions  
U. Kuppuswamy, S. Ananthasubramanian, Y. Wang, University of Pittsburgh; N. Balakrishnan, Indian Institute of Science; M. Ganapathiraju, University of Pittsburgh  
Board 26
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<td>M. Samwald, Medical University of Vienna; R. Freimuth, Mayo Clinic; R. Powers, Predictive Medicine, Inc.; J. Luciano, Rensselaer Polytechnic Institute; E. Prud’hommeaux, World Wide Web Consortium; R. Boyce, University of Pittsburgh; M. Marshall, Leiden University Medical Center; M. Dumontier, Carleton University</td>
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<td>AERSmine: A Phenome-pharmacome Web Datamine Based on the FDA’s Adverse Events Reporting System Data</td>
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<td>X. Sun, N. Tatonetti, Columbia University</td>
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<td>A. Surkis, A. McCrillis, R. McGowan, NYU School of Medicine; B. Schmidt, NYU College of Dentistry</td>
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<td>Modulating Cancer Progression from Leukoplakia via Bayesian Gene Networks</td>
<td>A. Villa, Brigham and Women’s Hospital and Harvard School of Dental Medicine; A. Zollanvari, A&amp;M Texas; S. Sonis, Brigham and Women’s Hospital - Division of Oral Medicine</td>
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<td>Terminology for a Centralized Biobank Information Management System</td>
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<td>Semantic Integration of Genotype-phenotype Resources</td>
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Integrated Gene Expression Probabilistic Models for Cancer Staging
A. Xia, MIT

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PharmGKB: Automating Pharmacogenomics Literature Curation
M. Whirl-Carrillo, D. Hewett, M. Woon, R. Altman, T. Klein, Stanford University

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Integrated Gene Expression Probabilistic Models for Cancer Staging
A. Xia, MIT

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Topic Modeling for Discovering Drug-related Adverse Events from Social Media
M. Xie, University of Arkansas at Little Rock; J. Bian, U. Topaloglu, University of Arkansas for Medical Sciences

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Integrated Sequence/Structure Analysis Associates Mycobacterium Tuberculosis (TB) Disordered Protein Regions with TB Drug Resistance
P. Zhang, Harvard Medical School; L. Zhang, Chinese Academy of Sciences; G. Alterovitz, Harvard Medical School

Board 57

Network-based Analysis Reveals Distinct Association Patterns in a Semantic MEDLINE-based Drug-disease-gene Network
Y. Zhang, C. Tao, Mayo Clinic; G. Jiang, A. Nair, J. Su, H. Liu, Mayo Clinic College of Medicine

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OHSU provides a detailed overview of biomedical and health informatics to those who work at the interface of healthcare and information technology (IT). It provides a broad understanding of the field from the vantage point of those who implement, lead, and develop IT solutions for improving health, healthcare, public health, and biomedical research. It provides up-to-date details on current events in the field, including the "meaningful use" of electronic health records specified by the Health Information Technology for Economic and Clinical Health (HITECH) Act of the American Recovery and Reinvestment Act (ARRA, also known as the economic stimulus package) of 2009. 46.5 CMEs available

To register: amia.org/education/10x10-courses
**How Can AMIA Best Support the Goals of Translational Bioinformatics and Clinical Research Informatics?**

As AMIA grows, the informatics tent grows larger to include the spectrum of domains from translational bioinformatics and clinical research informatics to applied clinical and operational informatics. Members throughout the community have expressed interest in a strategic vision that is rooted in the core research and science interests and also includes expanded outreach and services for applied clinical informaticians.

Join Gilad Kuperman, AMIA Board Chair, and Kevin M. Fickenscher, AMIA President and CEO, for an open Town Hall event with Q&A. The AMIA leadership is listening and welcomes full attendance at this session. Topics will include work of the Board of Directors, new and continuing initiatives, and an update on AMIA's strategic directions.

**ERIC HORVITZ, MD, PhD**  
Deputy Managing Director at Microsoft Research  
(See page 13 for description.)

The number of doctoral training programs in informatics increases every year, however not every doctoral candidate wishes to pursue a traditional career in academia. In addition, the knowledge and skills acquired through scientific training at the doctoral level can be valuable, even critical, for a number of career paths outside of academic research and teaching. This panel will present a diverse set of alternative career paths for which graduates of Informatics programs would be well suited, including patent law, research in industry, academic administration, and scientific journalism. Panelists will describe their own respective backgrounds and career paths, a day in the life in their current position, and how their training prepared them for their jobs. They will also touch on insights gained and lessons learned in exploring the professional landscape through non-traditional paths.
TBI-15: Papers/Podium Presentations - Electronic Health Record Data Mining
Room: Cyril Magnin II
Session Chair: Joshua Denny

Automated Detection of Systematic Off-label Drug Use in Free Text of Electronic Medical Records
K. Jung, P. LePendu, N. Shah, Stanford University

Scalable Data-driven Phenotypes via Unsupervised Feature Learning
T. Lasko, J. Denny, M. Levy, Vanderbilt University Medical Center

Evaluation Considerations for EHR-based Phenotyping Algorithms: A Case Study for Drug-induced Liver Injury
C. Overby, C. Weng, K. Haerian, A. Perotte, C. Friedman, G. Hripcsak, Columbia University

From EHRs to Linked Data: Representing and Mining Encounter Data for Clinical Expertise Evaluation
C. Torniai, S. Essaid, Oregon Health & Science University; C. Barnes, University of Florida; J. Hajagos, E. Bremer, Stony Brook University; J. Corson-Rikert, Cornell University; M. Haendel, Oregon Health & Sciences University

CRI-01: Papers/Podium Presentations - EMR Text Processing/Phenotyping 1
Room: Cyril Magnin III
Session Chair: Hua Xu

Learning Drug-drug Interactions from the Unstructured Text of Electronic Health Records
S. Iyer, P. LePendu, R. Harpaz, A. Bauer-Mehren, N. Shah, Stanford University

S. Meystre, University of Utah; O. Fernandez, Nuance Communications Inc.; B. South, VA Salt Lake City Health Care; S. Shen, M. Samore, University of Utah

Extracting Computational and Semantic Features from Portable Chest X-rays for Diagnosis of Acute Respiratory Distress Syndrome
H. Fan-Minogue, D. Maslove, Stanford University; P. Lamb, Biomedical Image Analysis Lab; J. Levitt, D. Paik, D. Rubin, Stanford University

Automated Tools for Phenotype Extraction from Medical Records
M. Yetisgen-Yildiz, C. Bejan, University of Washington; L. Vanderwende, Microsoft Research; F. Xia, H. Evans, M. Wurfel, University of Washington

CRI-02: Panel - Integrating Governance of Research Informatics and Healthcare IT across an Enterprise: Experiences from the Trenches
Room: Mission

P. Embi, The Ohio State University; U. Tachinardi, UW-Madison; Y. Lussier, University of Illinois at Chicago; J. Starren, Northwestern University; J. Silverstein, NorthShore University HealthSystem

Advances in health information technology and biomedical informatics have laid the groundwork for significant improvements in healthcare and biomedical research. For instance, Electronic Health Records can help improve the delivery of evidence-based care, enhance quality, and contribute to discoveries and evidence generation. Despite this promise, there are many challenges to achieving the vision and missions of our healthcare and research enterprises. Given the challenges inherent in doing so, institutions are increasingly moving to establish dedicated leadership and governance models charged with designing, deploying and leveraging various information resources to advance research and advanced care activities at AHCs. Some institutions have even created a new leadership position to oversee such activities, such as the Chief Research Information Officer. This panel will include research informatics leaders discussing their experiences from the proverbial trenches as they work to operationalize such cross-mission governance models. Panelists will start by providing an overview their respective positions and environments, discuss their experiences, and share lessons learned through their work at the intersection of clinical and translational research informatics and Health IT.
12:15 p.m. – 1:15 p.m. Lunch & Learn

Clinical Data Management for Adverse Events: tracking and reporting of events based on Common Toxicity Criteria and Graft-Versus-Host Disease grading standards—using mobile solutions from Velos.
(See page 15 YELLOW for description.)

1:30 p.m. – 3:00 p.m. Scientific Sessions

**TBI-16: Papers/Podium Presentations – TBI in Oncology**
Room: Cyril Magnin I
Session Chair: Yohan Lee

Qualitative and Quantitative Image-based Biomarkers of Therapeutic Response in Triple-negative Breast Cancer
D. Golden, Stanford University; J. Lipson, M. Telli, J. Ford, Stanford University Medical Center; D. Rubin, Stanford University

Platform for Personalized Oncology: Integrative Analyses Reveal Novel Molecular Signatures Associated with Colorectal Cancer Relapse
S. Madhavan, Georgetown University

Domain Landscapes of Somatic Mutations in Cancer
T. Peterson, D. Park, M. Kann, University of Maryland, Baltimore County

Database Integration of 2256 Publicly-available Samples of Breast Cancer Molecular and Clinical Data
C. Planey, A. Butte, Stanford University

**TBI-17: Papers/Podium Demonstrations – Pharmacogenomics Decision Support**
Room: Cyril Magnin II
Session Chair: Robert Freimuth

Providing Pharmacogenomics Clinical Decision Support Using Whole Genome Sequencing Data as Input
V. Huser, J. Cimino, National Institutes of Health, Clinical Center

Toward Semantic Modeling Pharmacogenomic Knowledge for Clinical and Translational Decision Support
R. Boyce, University of Pittsburgh; R. Freimuth, Mayo Clinic; K. Romagnoli, T. Pummer, H. Hochheiser, P. Empey, University of Pittsburgh

Development of a Scalable Pharmacogenomic Clinical Decision Support Service
V. Fusaro, Harvard Medical School; C. Brownstein, W. Wolf, C. Clinton, S. Savage, Children’s Hospital Boston; K. Mandl, Harvard Medical School; D. Margulies, S. Manzi, Boston Children’s Hospital

A Framework of Knowledge Integration and Discovery for Supporting Pharmacogenomics Target Predication of Adverse Drug Events: A Case Study of Drug-induced Long QT Syndrome
G. Jiang, C. Wang, Q. Zhu, Mayo Clinic College of Medicine; C. Chute, Mayo Clinic

**CRI-03: Silver Medallion CRI Poster Review**
Room: Mission

J. Cimino, National Institutes of Health Clinical Centers; W. Hammond, Duke Center for Health Informatics; M. Kahn, University of Colorado; K. Mandl, Boston Children’s Hospital
Silver Medallion CRI Poster Review is new this year and 120 posters are included in the CRI Poster Session. So many posters, so little time! This novel session will feature a selection of top posters as presented, contextualized, and discussed by some of the most distinguished researchers and AMIA members in the field of Clinical Research.

(See page 28 YELLOW for list of posters, listed alpha order by first author.)

### CRI-04: Papers/Podium Presentations - EMR Texting/Phenotyping 2
Room: Cyril Magnin III

**Session Chair:** Jorge Herskovic

- **How Many Patients are "Normal"? Only 1.55%**
  G. Weber, Beth Israel Deaconess Medical Center

- **Diagnostic Prediction of Von Willebrand Disease using Multiple Bleeding Phenomics Datasets**
  S. Mollah, The Rockefeller University

- **"Sitting on Pins and Needles": Characterization of Symptom Descriptions in Clinical Notes**
  T. Forbush, VA Salt Lake City Healthcare System; A. Gundlapalli, University of Utah; M. Palmer, VA Salt Lake City Health Care System; S. Shen, University of Utah; B. South, VA Salt Lake City Health Care; G. Divita, M. Carter, A. Redd, University of Utah; J. Butler, Veteran’s Affairs; M. Samore, University of Utah

- **Extracting Actionable Findings of Appendicitis from Radiology Reports Using Natural Language Processing**
  B. Rink, K. Roberts, S. Harabagiu, University of Texas at Dallas; R. Scheuermann, J. Craig Venter Institute; S. Toomay, T. Browning, T. Bosler, R. Peshock, U.T. Southwestern Medical Center

3:00 p.m. – 3:30 p.m.  **Coffee Break**  Room: Cyril Magnin Foyer

3:30 p.m. – 5:00 p.m.  **TBI Closing Session: Translational Bioinformatics**  Room: Cyril Magnin I/II

**Year in Review**

**RUSS ALTMAN, MD, PhD**

Professor of Bioengineering and Genetics, Stanford University

(See page 13 YELLOW for description.)

6:00 p.m. – 7:30 p.m.  **Natural Language Processing Working Group Business Meeting**  Room: Balboa

6:00 p.m. – 8:00 p.m.  **i2b2 Academic Users’ Group (AUG) Workshop (Affiliate Event)**  Room: Mission

6:00 p.m. – 9:30 p.m.  **The Ohio State University Red & Grey Reception (Affiliate Event)**  Room: Cyril Magnin III