Knowledge Mining and Bioinformatics Tools to Advance Personalized Diagnostics and Therapeutics

February 4-5, 2012
Florence Italy

Organized by:
The U.S.-Israel Science and Technology Foundation
The Technion – Israel Institute of Technology
Florida International University
University of Maryland – Baltimore County

Generous Support Provided by:
U.S. National Science Foundation
PROJECT REPORT

USISTF-organized Workshop Brings Together Medical and Computer Scientists from USA, Europe, Israel, and Middle East to Advance Knowledge Mining and Bioinformatics Tools for Personalized Diagnostics and Therapeutics

“We have today a better cooperation between the Bio-Medical and Genetic Research and the advanced electronic Medical Health records, but we need more workshops of this kind in order to bring again computer scientists and clinicians to come together and lay the groundwork for the future of medicine by providing the relevant information to the general practitioner in front of the patient.”
- Uzia Galil, Workshop Participant and Founder of the Galil Center for Medical Informatics, Telemedicine and Personalized Medicine at the Technion – Israel Institute of Technology

The Need:

The world has seen the human genome fully decoded by an international team of scientists after more than a decade of work to being available to scientists within days or hours. The question now is how will we use the wealth of information available to us through our newly understood genomic data and, further, given our massive computing power, can we merge this information with all of a given patient’s health data, compared against that of similar cases, in order to better diagnose and offer therapeutics?

Program Conveners:

The multi-disciplinary workshop was imagined and convened by a group of interdisciplinary experts representing the United States and Israel and implemented adjunct to the International Congress on Personalized Medicine (UPCP).

- The U.S.-Israel Science and Technology Foundation
  - Ms. Ann Liebschutz, Organizational Chair
  - Ms. Eve Copeland, Workshop Logistics Manager

- Florida International University
  - Dr. Naphtali Rishe, Workshop Chair and National Science Foundation Principle Investigator

- University of Maryland, Baltimore County
  - Dr. Yelena Yesha, Program Chair

- Technion – Israel Institute of Technology
  - Dr. Eddy Karnieli, Up Close and Personalized Medicine Congress General Chair and Israel Chair

Workshop Participants:

The workshop brought together a group of 40 medical and computer scientists representing the United States, Israel, Palestinian Territories, Canada, Italy, Turkey, the Netherlands and Germany. Institutions such as: Technion, Ben Gurion University, IBM Canada, University of Florence, the Max Planck Institute, University of Maryland Medical School, Emory University, Duke University, Florida Atlantic University, Al-Quds University, Groningen University Medical Centre, Rockefeller Center for e-Health, Kigali Institute of Science and
Technology, Akdeniz University, Israel National Health Services (Clalit), Texas Oncology – Baylor Charles A. Sammons Cancer Center and the University of Arizona Medical School.

The uniqueness of this workshop is embodied by the wide diversity of workshop participants all of whom came from interdisciplinary backgrounds. Computer scientists trained to develop software and hardware to analyze ‘big data’ sat with genomic data researchers and medical clinicians to discuss the future of personalized diagnostics and hash out exactly what steps the group might need to take to address all aspects of future needs. Appendix A of this report offers a detailed participant directory.

**Scientific Panels and Workshop Next Steps:**

The workshop included four scientific sessions focusing on all aspects of developing and implementing big data driven personalized diagnostics and therapeutics. The first panel, “Delivering Personalized Medicine: Vision, Policy and Challenges” looked at the ways that companies such as IBM are creating and utilizing personal web platforms to drive bioinformatics utilization in diagnostics. It also examined the policy considerations that would protect patient privacy while enabling researchers to utilize available data and the next steps to integrate privacy protection into available systems. The international perspective was touched on by scientists from Al-Quds University who described both the opportunities for expanding personalized medicine techniques at the academic level and the cultural challenges of introducing new therapeutic techniques in the West Bank. Intellectual Property challenges were also examined through the lens of a the high courts considering who owns the IP after participating in clinical trials utilizing genomic data.

The technology panel titled “Big Data’ Challenges” considered both the software necessary and currently available to analyze what we call the ‘big data’ generated by patient’s natural health records, genomic sequences and metabolic data and new ways of assessing it such as mobile based home healthcare solutions. Scientists also presented “High Throughput Sequencing Platforms and Genomics Structural Variants” which offered novel ways of analyzing and understanding the genomic data being generated by researchers. Researchers actually producing the analysis of genomic data also evaluated the state of quality assessment and exploratory analysis of ‘big data’ to provide a state of the industry discussion about what they can do today and what they hope to be able to tomorrow.

The prediction panel “Personalized Medicine: The Science of Personalized Medicine” honed in on the ways that the tools being developed by computer scientists can realistically be used today and the bedside for treatment and prediction. Scientists in the prediction panel presented their work refining models and techniques for using personalized medicine data such as second generation sequencing reads to map the structural variations, SVs, single-nucleotide polymorphisms, and SNPs to attempt to predict more refined outcomes in family groups. Other groups discussed the models and challenges of building a prediction model based on clinical and genomic datasets which include not only genomic information, but basic personal data and secondary lifestyle habits such as fat intake, fiber intake, glycemic load to try to create clusters to drive prediction of diseases such as diabetes. The clusters yield interesting hints for potentially relevant combinations of clinical-risk factors that would
certainly benefic researcher for analyzing case-control groups, gain more insight into the genetic behaviors and generate biological hypotheses.

Finally, the medical panel, “Personalized Medicine: Challenges in Delivery” explored the ways that doctors are using the big data garnered from new knowledge mining techniques to improve their diagnostics and therapeutics at the bedside. Researchers shared their work ranging from utilizing nano-medical techniques based on personalized data to utilize a diagnostic imaging aid to target moiety and develop agents that can overcome drug resistance in the context of a targeted chemotherapy regime. Researchers also shared work looking at ways to use bioinformatic data to target MBP inhibitors that play a strong role in drug resistance, the major challenge in cancer treatment.

The workshop, fully moderated by Dr. Ron Ribitzky, the workshop concluded by reaching multi-disciplinary and multi-national broad-based consensus on new directions for knowledge mining and bioinformatics tools to impact patient care; as well as strategic, proactive, and preventive health and wellness decisions here and now. A multi-faceted, grand-challenge undertaking, the highlights included call-to-action for technological breakthroughs to fill the growing ‘translational white spaces’ among the many scientific and clinical disciplines throughout the personalized medicine cycle up to end-user clinicians, patients, and consumers; business models innovation to accelerate the reduction of new discoveries along that cycle to practice; and policies that facilitate both. In terms of partnerships, scientists from Al-Quds University are already interested in creating new programs in the university’s medical school that integrate personalized concepts and in developing a bi-lateral program to bring Al-Quds students to Florida International University to study computer science techniques to enhance their ‘big data’ analysis capabilities. Collaborations are already coming together around a project led by UMBC to develop a platform for clinician analysis of patient data at the bedside and new collaborators will be joining the project spurred by their discussions at the workshop. The group is preparing to deliver the scientific proceedings of the workshop to the National Science Foundation and broader community. These proceedings are expected to be published as Springer Verlag Lecture Notes in Computer Science.

Uzia Galil, the founder of the Galil Center for Medical Informatics, Telemedicine and Personalized Medicine at the Technion – Israel Institute of Technology, had this to say about the workshop which he participated in, “we have today a better cooperation between the Bio-Medical and Genetic Research and the advanced electronic Medical Health records, but we need more workshops of this kind in order to bring again computer scientists and clinicians to come together and lay the groundwork for the future of medicine by providing the relevant information to the general practitioner in front of the patient."
Appendix A.

“Knowledge Mining and Bioinformatics Tools to Advance Personalized Diagnostics and Therapeutics”

Panelists and Participants


- **Dr. Joanna Ng**, Head of Research at IBM Canada Software Laboratories, Center for Advanced Studies. Senior Technical Staff Member, IBM Software Group.
- **Dr. Anupam Joshi**, Oros Family Professor of Computer Science and Electrical Engineering, University of Maryland, Baltimore County
- **Dr. Karuna Joshi**, PHD Candidate, Computer Science in the field of Cloud Based Services and Distributed Web Systems from University of Maryland, Baltimore County
- **Dr. Jessica Nadler**, Manager, Deloitte
- **Dr. Hussein Hallak**, Assistant Professor, Faculty Member, College of Pharmacy, Al-Quds University
- **Ms. Ann Liebschutz**, Executive Director, U.S.-Israel Science and Technology Foundation
- **Dr. Hasan Salah Dweik**, Executive Vice President, Al-Quds University, Director of Science Discovery Center and Mathematics Museum

*Technology Panel*: “Big Data’ Challenges”

- **Dr. Naphtali Rishe**, Professor of Computer Science, Florida International University and Director, NSF Industry/University Cooperative Research Center for Advanced Knowledge Enablement (I/UCRC-CAKE)
- **Dr. Vered Caspi**, head of the Bioinformatics Core Facility at the National Institute of Biotechnology in the Negev, at Ben-Gurion University
- **Dr. Borko Furht**, Professor and Chairman of the Department of Electrical and Computer Engineering and Computer Science at Florida Atlantic University. Director of Industry/University Cooperative Research Center on Advanced Knowledge Enablement
- **Dr. Stefano Ruffo**, Professor, Faculty of Engineering, University of Florence
- **Dr. Alberto Magi**, Faculty of Medicine, University of Florence


- **Dr. Yelena Yesha**, Professor, Computer Science, University of Maryland Baltimore County
Dr. Christopher Newgard, Director of the Sarah W. Stedman Nutrition and Metabolism Center and the W. David and Sarah W. Stedman Distinguished Professor of Pharmacology and Cancer Biology at the Duke University Medical Center

Dr. Derek LeRoith, Diabetes and Metabolism Clinical Research Center of Excellence, Legacy Heritage Clinical Research Institute at Rambam (LHCRIR)

Mr. Abhishek Narain Singh, ICT Associate at University Medical Centre, Groningen, The Netherlands

Dr. Yaacov Yesha, Professor, Computer Science and Engineering, University of Maryland Baltimore County

Medical Panel: “Personalized Medicine: Challenges in Delivery”

Dr. Eddy Karnieli, Director, Institute of Endocrinology, Diabetes and Metabolism, RAMBAM Medical Center

Dr. Joel Saltz, Chair of the Department of Biomedical Informatics and Director of the Center for Comprehensive Informatics at Emory University

Dr. Eliot Siegel, is Professor and Vice Chairman of Radiology for the University of Maryland School of Medicine Department of Diagnostic Radiology and Nuclear Medicine and Chief Imaging for the VA Maryland Healthcare System

Dr. Ron Ribitzky, CEO, R&D Ribitzky, Distinguished Visiting Professor, Rockefeller Center for e-Health, Kigali Institute of Science and Technology, Rwanda

Dr. Yousef Najajreh, Dean of Scientific Research/Al-Quds University, Head of the Projects Department/Planning and Development Office/Al Quds University/ Head of the Anticancer Drugs Research Lab

Dr. Alan Shuldiner, John L. Whitehurst Professor of Medicine and Associate Dean for Personalized Medicine at the University of Maryland School of Medicine. Directs UMSOM Program in Personalized and Genomic Medicine, serves as Head of the Division of Endocrinology, Diabetes and Nutrition in the Department of Medicine and is an Investigator at the Baltimore Veterans Administration Geriatrics Research and Education Clinical Center

Dr. Yoav Livney, Senior Lecturer, Laboratory of Biopolymers and Food-Nanotechnology, Department of Biotechnology and Food Engineering, Technion

Participants

Dr. Alexander Kuhn, Post-Doctoral Researcher, Molecular Genetics, Max Planck Institute

Prof. Mehmet Akif Kilic, Akdeniz University, Turkey

Dr. Sean Szeja, College of Medicine, University of Arizona, USA

Dr. Efrat Recanati, Clalit Health Services, Israel

Dr. Christoph Wierling, Max Planck Institute for Molecular Genetics, Germany

Dr. Robert Mennel, Oncologist, Texas Oncology-Baylor Charles A. Sammons Cancer Center

Dr. Andreas Lininger
Dr. Shmuel Klang, Clalit Health Services, Israel

Dr. Nicky Lieberman, Clalit Health Services, Israel

Dr. Ondrej Topolman, Czech Republic

Mr. Uzia Galil, Founder, Galil Center for Medical Informatics, Telehealth and Personalized Medicine, Israel

Dr. Tajana Lucic, United States

Dr. Ariel Miller, Technion, Israel

Ms. Elva Ngai-Yu Lei, PHD Candidate, Chemistry, City University of Hong Kong
Appendix B. Official Workshop Agenda

“Knowledge Mining and Bioinformatics Tools to Advance Personalized Diagnostics and Therapeutics”

Scientific Agenda

February 4th, 2012

<table>
<thead>
<tr>
<th>Time</th>
<th>Program</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>8:00 AM – 8:30 AM</td>
<td>Participant Welcome and Registration</td>
<td>Hall C</td>
</tr>
<tr>
<td>8:30 AM – 10:30 AM</td>
<td>Joint Session with Up Close and Personalized Medicine Congress: Genomics</td>
<td>Hall A</td>
</tr>
<tr>
<td>11:00 AM – 1:00 PM</td>
<td>Policy Panel: “Delivering Personalized Medicine: Vision, Policy and Challenges”</td>
<td>Hall C</td>
</tr>
<tr>
<td></td>
<td>Dr. Joanna Ng</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Karuna Joshi</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Hussein Hallak</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Hasan Dweik</td>
<td></td>
</tr>
<tr>
<td>1:00 PM – 2:00 PM</td>
<td>Lunch</td>
<td></td>
</tr>
<tr>
<td>2:00 PM – 4:00 PM</td>
<td>Technology Panel: ‘‘Big Data’ Challenges’’</td>
<td>Hall C</td>
</tr>
<tr>
<td></td>
<td>Dr. Naphtali Rishe</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Vered Caspi</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Borko Furht</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Stefano Ruffo</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Alberto Magi</td>
<td></td>
</tr>
</tbody>
</table>

February 5th, 2012

<table>
<thead>
<tr>
<th>Time</th>
<th>Program</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>9:00 AM – 10:30 AM</td>
<td>Prediction Panel: “Prediction: The Science of Personalized Medicine”</td>
<td>Hall C</td>
</tr>
<tr>
<td></td>
<td>Dr. Yelena Yesha</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Derek LeRoith</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Mr. Abhishek Narain Singh</td>
<td></td>
</tr>
<tr>
<td>10:45 AM – 12:30 PM</td>
<td>Medical Panel: “Personalized Medicine: Challenges in Delivery”</td>
<td>Hall C</td>
</tr>
<tr>
<td></td>
<td>Dr. Joel Saltz</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Ron Ribitsky</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Dr. Eddy Karnieli</td>
<td></td>
</tr>
<tr>
<td>12:30 PM – 1:30 PM</td>
<td>Lunch: Speaker Dr. Joel Saltz “Role of Clinical Informatics in Personalized Medicine”</td>
<td>Hilton Metropole</td>
</tr>
<tr>
<td>1:30 PM – 4:30 PM</td>
<td>Workshop: De Bono Innovation Exercise and Next Step Discussion</td>
<td>Hall C</td>
</tr>
</tbody>
</table>