My surgeon Dr. Shawna Willey walked into the patient exam room where I waited nervously. I first saw her thumbs up before her beaming face. I could breathe again!

My friends and I who recently turned 40 and starting our baseline mammograms can’t help but wonder about the lack of consensus on optimal cancer screening strategies, target populations, its benefits and harms. My colleague Dr. Jeanne Mandleblatt and her team have studied breast screening strategies for decades and have shown that biennial screening from ages 50-74 achieves a median 25.8% breast cancer mortality reduction whereas annual screening from ages 40-74 reduces mortality an additional 12% but introduces very high false positive rates. Many women and their families are subject to extreme anxiety due to the sheer number of repeat mammograms, false-positives, benign biopsies and in 7% of the cases an over diagnosis.

My experience and of my friends with breast cancer screening are raising many questions. How can we better predict the target risk population who must undergo screening early and often? Would this decision-making process consider risk factors, lifestyle, and patient preferences? How

ICBI Annual Symposium Registration
Now Open!

Register today!

The deadline for abstract submission is Sep 15th, 2017.

Early Bird Registration ends on Sep 7th, 2017

ICBI’s 6th Annual Biomedical Informatics Symposium will be held on Friday, October 27th, 2016 at the Georgetown University Conference Center and Hotel. This one-day event will showcase exciting sessions on cancer networks, machine learning, artificial intelligence and other emerging technologies in healthcare and precision medicine.

Dr. Patricia Flatley Brennan, Director of the National Library of Medicine and Interim Associate NIH Director for Data Science will deliver the keynote address.

Click HERE for more information on the event and abstract submission.

A summary of last year’s symposium including poster award winners is available here.
often are patients with a diagnosis of a benign breast condition on a stereotactic core needle biopsy upgraded to a non-benign diagnosis on an excisional biopsy which requires full sedation and surgery? What was the care journey like for other patients like me – Asian female, healthy, no family history? How many in the US and globally have access to the excellent care and follow-up that I was privileged to receive from Dr. Willey and her expert team?

Touted as the fourth industrial revolution, Artificial Intelligence is poised to empower clinicians, patients and researchers in answering these questions. What is AI? The term was coined by Dartmouth professor Dr. John McCarthy in 1956 and defined as “the science and engineering of making intelligent machines, especially intelligent computer programs.” Applications of AI in medicine have been limited by the complexity of highly cognitive processes such as making a medical diagnosis or selecting a treatment which require integration of thousands of datasets with millions of variables and multiple interactions between these variables. It takes years to collect, organize and publish practice changing results such as Jeanne’s screening study. What if we could use data that we routinely collect during the care process and effectively use AI to assist clinicians in real-time to make informed treatment decisions?

Interested in learning more about AI in Biomedicine? Want to engage with expert scientists and product developers in AI? Register for Georgetown’s Big Data in Biomedicine symposium on October 27th!

Companies like Google and Amazon are betting big on this. Jeff Bezos wrote “…it is hard to overstate how big of an impact AI will have on society over the next 20 years”; Google’s Sundar Pichai, when asked recently about the next big thing at Google responded “I can’t quite tell exactly but advances in AI and machine learning, we are making a big bet on that and this will bring a difference in many many fields”.

We cannot have a conversation about AI in medicine without discussing IBM Watson, the supercomputer that sifted through 20 million cancer research papers, and conducted a differential diagnosis on a difficult to treat leukemia patient in 10 minutes by combining genomic data with the power of cognitive computing. One concern that informaticians including my informatics mentor Dr. Bill Hersh have raised is that the publicity around Watson

**Cannabis Genome Research Program**

Teewinot has entered into an ambitious Cannabis genome research program in collaboration with ICBI and Seach Ltd. The goal of this project is to decipher the Cannabis Genome and identify new methods of manufacturing cannabinoid-based therapies using proprietary biosynthetic methods. [Read more](#)

**Clingen Somatic Cancer Variant Curation**

The Clingen Cancer Somatic Workgroup co-led by Dr. Subha Madhavan and Dr. Sashi Kulkarni, Baylor College of Medicine in collaboration and consensus with various expert groups published standards for the curation of somatic variants in cancer and interpretation of their clinical actionability (MVLD framework).

Biocurators at ICBI and the Clingen community are currently adopting this framework to collect and interpret somatic variants in cancer within the Clivic system. We will be presenting a poster on our work at the ‘Curating the Clinical Genome’ conference on June 28th 2017 in Washington D.C.

**mHealth – Gulf War Complementary and Alternative Medicine (GWCAM)**

In 2015, ICBI developed a mobile health application for a DoD funded project to help the VA evaluate the efficacy of a Complementary and Alternative Medicine (CAM) program for sleep, health functioning and quality of life intervention. The mobile app is being used for data collection and monitoring of specific symptoms such as fatigue, pain, cognitive deficit and sleep disturbance to enable increased health related functioning through the CAM program. This study is now recruiting its fourth cohort of patients. We are currently repurposing this mobile application for use in additional clinical trials at the VA.

**New REDCap version 6.18.1 is now available at Georgetown University**

Please visit the REDCap Georgetown homepage for the list of new features,
has mostly been from news articles and press releases, primarily from researchers at IBM and call for a more scientific analysis, not n-of-one case reports, of its abilities in clinical decision making. Systems like Watson will benefit from systematic expert knowledge input to guide the cognitive computing processes in navigating the complex medical pathways.

While still early, AI is already starting to make important contributions to Medicine says AI professor at MIT and a recent breast cancer survivor, Dr. Regina Barzilay. She and her team are asking all the right questions of data – “can we apply the sophisticated algorithms we use to predict customer’s shoe-buying habits to adjust treatments for cancer patients?” “Can computers detect signs of breast cancer or even pre-malignancy earlier than humans are currently capable of?” And the Holy Grail – “Can we use the huge quantities of data from smart toothbrushes, wearables, genomic sequencing, medical records to get to the first and right treatment?”

What next?

In the last decade, big data in biomedicine has focused on collecting (e.g. through mobile and other IoT) and organizing (e.g. cloud computing) information but all signs point in one direction for the next decade – real world applications of AI. We will witness the development of expert systems, question-answering systems and deep learning methods that begin to address complex real world problems in medicine. These will augment, not replace, human expertise. Winners will find ways to rapidly and accurately integrate human input with computational output. Usability of these tools by end users and human factors will be key.

While a true tech automation enthusiast at heart and practice, I will never forget Dr. Willey’s kind and soft words as she clearly explained my pathology report. She also carefully noted in my medical record the rare chlorohexidine pre-op antiseptic agent hypersensitivity that I had developed post anesthetic induction.

One more data point!

Let’s continue the conversation:

- subha.madhavan@georgetown.edu
- Twitter @subhamadhavan

REDCap policies and service request form.

**edX MOOC: Demystifying Biomedical Big Data: A User’s Guide.**

ICBI is proud to announce that we have completed our very first session of the Massive Open Online Course (MOOC) on edX. The goal of this course is to “demystify” the process of analyzing biomedical big data through a series of lectures and online hands-on training sessions and demos. For more information and to enroll, please visit the course webpage.

**ICBI Blog**

Dr. Simina Boca wrote a series of posts on the ICBI blog, called “An injection of optimism into biomedical research”

- Newborn screening success!
- Improved management for two textbook diseases
- From deadly to chronic.
- Cancer prevention and early detection
- Millions of babies

**New Appointments**

**ICBI is delighted to welcome new team members, collaborators and students**

Dr. Matthew McCoy joined ICBI as a new faculty member in November 2016. Dr. McCoy's current research aims to understand how genomic mutations alter protein function, and how functional changes impact the emergent behavior in biological systems.

Dr. Keeshaloy Thompson is a Translational Biomedical Science (TBS) program scholar at GU. She is currently collaborating with scientists at ICBI to enable her research on the metabolism of small molecules that interact with nicotinic receptors in the brain and using molecular
ICBI Seminars

Once a month, ICBI invites leading researchers from around the world to give a seminar to the Georgetown biomedical informatics community. Keep an eye out for emails from Dr. Simina Boca for announcements on upcoming seminars. Below are some recent seminars:

"Informatics programs in the Surveillance, Epidemiology and End Results (SEER) cancer registries" by Lynne Penberthy, MD, NCI.

"Statistics and Visualization for Metagenomic Data Analysis" by Hector Corrada Bravo, Ph.D, UMD

"Development of Pharmacodynamic Biomarkers in Duchenne Muscular Dystrophy" by Yetrib Hathout, Ph.D, Children’s National Health System and GWU.

"Optimizing Molecularly Targeted Cancer Therapies with Imaging" by Sridhar Nimmagadda, Ph.D of Johns Hopkins Medicine.

“The Effective use of Epigenomic Data to Better Understand Complex Diseases” by Mikhail Dozmorov, Ph.D Virginia Commonwealth University Medical Center

“Medical Genomics Japan Database: Introduction of Global Picture and Somatic Cancer Group” by Norihiro Kato, Ph.D (National Center for Global Health and Medicine NCGM Japan) and Katsuya Tsuchihara, PhD, M.D (National Cancer Center Japan).

“Microbiota of the lung and oral cavity and their relation to exposures” by Dr. Guoquin Yu, Ph.D, NCI

“Modeling of Inter- and Intra- Tumor Heterogeneity” by Dr. Simona Cristea, Postdoctoral fellow at Harvard University.

ICBI Social

Last month our team went on a day long sailing retreat in Annapolis. It was a gorgeous sunny day, perfect weather for sailing and team building!

ICBI Interns: Since last fall, ICBI faculty members have mentored Masters students, Mustafa Albahrani, Yi Chen, Stephanie Huang, Yulin Shi, Muzu Li and Ushna Ahmed from the Biochemistry and Biostatistics programs at GU.

We are excited to welcome our new summer interns this month who will work on a variety of multi-omics data analysis, software development and somatic variant curation projects.

Recent Publications


Chen, Cong, Deng, Qiqi, He, Linchen, Mehrotra, Devan V, Rubin, Eric H, and Beckman, Robert A. “How many tumor
ICBI in the Community

- **Russian Radiobiological Repository for Human Tissues (RRRHT)** In the month of September (2016) and again in March (2017), our Director of Health Operations and Technology Adil Alaoui traveled with a team of scientists to Ozersk, Russia for the RHTR. This unique biospecimen repository was established for the collection and storage of biological material which could be used in research on biological radiation effects.

- ICBI’s research work led by Dr. Yuriy Gusev on infiltrating lymphocytes with Oncologist Dr. Aiwu Ruth He and team was showcased in the Ruesch Center Annual Report.

*We have had a busy spring 2017. ICBI members participated in the following recent conferences, events and meetings.*

**Posters**

**Curating the Clinical Genome (2017)**

“Cancer Variant Curation for Clinical and Public Use: Disseminating Minimum Variant Level Data (MVLD) through Collaboration and Curation” - Deborah I. Ritter et al.

“MACE2K: Molecular and Clinical Extraction to Knowledge, a tool for Precision Medicine” - Shruti Rao et al.

“Combining Protein and Genome Annotations for Deciphering Functional Effects of Variation” – Peter McGarvey et al.

**AACR Annual Meeting (2017)**

The Georgetown Database of Cancer (G-DOC): A web-based data sharing platform for precision medicine *Krithika Bhuvaneshwar et al.*


RNaseq analysis of infiltrating immune cells in liver cancer. *Krithika Bhuvaneshwar et al.*

**Talks**

**AMIA 2017 Joint Summits on Translational Science (San Francisco)**

Panel speaker – Making Precision Oncology Data More Usable for Research and Care, Subha Madhavan

Panel speaker – Information Technology Powering Cancer Research for Discovery and Novel Hypothesis Generation, Subha Madhavan

Applying Protein Functional Annotation to Clinical Interpretation of Genomic Variants, Peter McGarvey.

A Human Factors Approach to Making Complex Cancer Precision Medicine Data Clinically Actionable, Vishakha Sharma

**10th EAI International Conference on Bio-inspired Information and Communications Technologies, 2017 (Hoboken, New Jersey)**

An Eye-tracking Study to Enhance Usability of Big Data in Cancer Precision Medicine, Vishakha Sharma
Variant analysis of LY6 genes in TCGA ovarian cancer. Krithika Bhuvaneshwar et al.

AMIA 2017 Joint Summits on Translational Science (San Francisco)

“The Georgetown Database of Cancer (G-DOC): A Web-Based Data Sharing Platform for Precision Medicine.” Krithika Bhuvaneshwar et al.


“MACE2K: Molecular and Clinical Extraction to Knowledge, a tool for Precision Medicine” - Shruti Rao et al.

ICBI Consultation and Support

We provide consultation and support to answer your research questions, help choose the most insightful analyses for your studies and run your data through our bioinformatics pipelines.

For more information on collaboration please contact us by completing the ICBI Service Request Form

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