

|       |  | Conference Day 1 – Wednesday April 26, 2017   |  |  |
|-------|--|---|--|--|
|       | OPENING KEYNOTE PLENARY  |   |  |  |
| 08:45 | Producers Opening Remarks Edward Glanville, Project Director, Terrapinn  |   |  |  |
| 08:50 | Opening remarks Chair: Atul Butte, Director, Institute for Computational Health Sciences, University of California, San Francisco  |   |  |  |
| 09:00 | Bringing big data and genomics to unlock cures for rare diseases     Connecting millions of data points to deliver ground breaking healthcare     Data is frozen knowledge. It's up to us to bring the heat to melt it     Investing in data  Atul Butte, Director, Institute for Computational Health Sciences, University of California, San Francisco.  |   |  |  |
| 09:20 | <ul> <li>Atul Butte, Director, Institute for Computational Health Sciences, University of California, San Francisco</li> <li>"Augmenting Human Insights with AI in Life Science"         <ul> <li>Advanced analytics, machine learning and artificial intelligence (AI) have emerged as powerful tools in healthcare and life science.</li> <li>Synthesizing diverse data is transforming the way we design experiments and investigate new therapies.</li> <li>Exploring the power of AI technologies &amp; its primary impact to augment human capabilities.</li> </ul> </li> <li>Bob Rogers PHD Chief Healthcare Data Scientist, Intel</li> </ul> |   |  |  |
| 09:50 | Elements of MVP (Million Veterans Project), where we want to go in the future, and our strategy to transform genomic efforts into the clinic.  Building one of the world's largest medical databases by safely collecting blood samples and health information from one million Veteran volunteers.  How to manipulate one of the largest genomic data sets in the world  Future use of MVP data to enhance the health of veterans  Ronald Przygodzki, Director, Genomic Medicine Implementation, U.S. Department of Veterans Affairs  |   |  |  |
| 10:10 | Speed networking and morning re  | efreshments   |  |  |
| 10:45 | GENOMICS AND HEALTH  | PRECISION<br>MEDICINE   | ARTIFICIAL<br>INTELLIGENCE   |  |
|       | Chair: Manuel Corpas, Scientific Lead, Repositive  | Chair: Paul W. Glimcher, Ph.D, Director, The HUMAN Project, Director, Institute for the Interdisciplinary Study of Decision Making, Julius Silver Professor of Neural Science, Economics, and Psychology, NYU | Chair: Alex Zhavoronkov,<br>(CSO, The Biogerontology<br>Research Foundation) CEO,<br>InSilico Medicine Inc |  |

| 10:50 | Facilitating a culture of |
|-------|---------------------------|
|       | responsible and effective |
|       | sharing of genome data    |

- Every disease is a rare disease at the molecular level
- Researchers will not have access to enough molecular test results for any rare disease without international data sharing
- Those of us involved in the Global Alliance for Genomics and Health are building successful mechanisms for international data sharing

**David Haussler,** Distinguished Professor of Biomolecular Engineering, **U.C.S.C.** 

### Genome Asia, sequencing 100,000 genomes across the Asian population

- Despite being >40% of the world's population - are significantly underrepresented in current genomic studies and reference genome databases even though the unique genetic diversity prevalent in South and East Asia provides a valuable source of clinical insights
- Developing a commitment to open information
- Understand biology of disease and enable new therapeutic options which will have global impact.

Stephan C. Schuster, Research Director, SCELSE, Professor at Nanyang Technological University, Singapore

#### Drug design at genomic scale

- We're remapping the entire chemical space of pharmaceuticals over the full human proteom.
- A conversion matrix that will be the foundation of new predictive and design models for highly selective, new therapeutics, and a new view on the biology of those.

**Riccardo Sabatini,**Co-Founder, Chief Data Officer, **Orionis** 

#### 11:10 PANEL DISCUSSION: Collaborative Scientific

Collaborative Scientific Innovation and Translational Medicine

- Christina Waters, CEO, RARE
- David Haussler,
   Distinguished Professor
   of Biomolecular
   Engineering, U.C.S.C.
- David Smith, Professor of Laboratory Medicine and Pathology Chairman of the Technology Assessment Group Center for Individualized Medicine, Mayo Clinic
- James Mills Barbeau,
   Assoc. Professor, Brown
   University Alpert
   Medical School,
   Director of Laboratory
   Medicine, Lifespan
   Academic Medical
   Center
- Scott Marshall Brandon Executive Director
   OpenMed Access

PANEL DISCUSSION: The Future of 'Omics: Mapping the Phenome and Why It Matters

- Paul W. Glimcher, Ph.D,
   Director, The HUMAN Project,
   Director, Institute for the
   Interdisciplinary Study of
   Decision Making, Julius Silver
   Professor of Neural Science,
   Economics, and Psychology,
   NYU
- Hannah Bayer, Chief Scientist, The HUMAN Project, Research Associate Professor of Decision Sciences, Institute for the Interdisciplinary Study of Decision Making, NYU
- Aristides Patrinos, Chief
   Scientist and Research
   Director, NOVIM
   Programs and Policy Adviser,
   Synthetic Genomics Inc. (SGI)
   Co-Founder, Joint Genome
   Institute of the Human
   Genome Project, U.S. Dept. of
   Energy
- Trevor Hawkins, Independent Board Member and Strategy Advisor, Past Director, Joint Genome Institute of the Human Genome Project, U.S. Dept. of Energy

#### MERCK PANEL

What problems in the drug discovery/development arena is most ripe for the application of AI/ML to have a large impact?

- What would a first rate approach to this problem look like in terms of the data, computing resources, machine learning algorithms, and domain knowledge needed?
- What is the best way to build out an AI/ML capability in a way that could tackle not only this problem but the variety of future opportunities that are coming?

Chair: Alex Zhavoronkov, (CSO, The Biogerontology Research Foundation) CEO, InSilico Medicine Inc

Mathai Mammen, Senior Vice President of Cardiovascular Diseases, Metabolic diseases, Immunology and Oncology, Merck

**Jeff Evelhoch,** VP Research Science and Translational Biomarkers, **Merck** 

**Devan Mehrotra,** AVP, Biostatistics, Early Development Statistics, **Merck** 

|       |   |  | Matt Tudor, Principal Scientist, Chemistry, Informatics, Merck  Carol Rohl, Exec. Dir, IT Account Management, Scientific Information Management, Merck  Dan Holder, Exec. Dir, Biometrics Research, Merck |
|-------|---|--|---|
|       | Interactive Roundtables   |  |   |
|       | GENOMICS AND HEALTH   | PRECISION MEDICINE   | ARTIFICIAL INTELLIGENCE   |
| 11:50 | Roundtable: Cleaning E-Health records and raw data. Inventing the car before the wheel. Scott Kahn, Ex CIO VP Informatics, Illumina   | Roundtable: Translation of NGS to the Clinic Christina Waters, CEO, RARE David Smith, Professor of Laboratory Medicine and Pathology Chairman of the Technology Assessment Group Center for Individualized Medicine, Mayo Clinic Catherine Brownstein, Scientific Director, Manton Center for Orphan Disease Research  | Roundtable: How to implement Al tools Thomas Clozel, co-founder, OWKIN  |
|       | Roundtable: American Heart Association Precision Medicine Platforms and funding opportunities  Jennifer Hall, President, American Heart Association Chris Riley,Research Mgr - Institute for Precision CV Medicine, American Heart Association Patrick Wayte,SVP, Center for Health Technology & Innovation, American Heart Association | Roundtable: Translating genomic studies in cancer into focused monitoring assays: CML, CLL, BTK and Beyond -Datamining to delivery: What makes a useful clinical molecular assay -Strategies for getting doctors to use the assay -Planning a path to reimbursement from the start.  Daniel Jones, Professor and Vice Chair, Division of the Molecular Pathology, Ohio State University Comprehensive Cancer Center Nikole Kimes, Senior Manager of Drug Safety and Public Health, Gilead Sciences | Roundtable: Developing innovative Al solutions for clinical research  Mark A. DePristo, Head of deep learning for genetics and genomics, Google Inc   |
|       | Roundtable: IT Infrastructure and HPC in Genomics Nicholas Marko, Founding board member of the International Society for Chief Data Officers, Director of Neurosurgical Oncology, Geisinger Health System   | Roundtable: How do we drive epigenetics into the clinical practice?  Jeffrey Bhasin, Clinical Epigenomics Leader, Cleveland Clinic   | Roundtable: The future of AI in pharmaceutical development  Philip Nelson, Director, Software Engineering, Google Inc   |

|       | Pablo Roman-Garcia, Industry Officer, ELIXIR  Roundtable How to drive forwards innovation in precision medicine through collaboration.  Scott Marshall Brandon Executive Director OpenMed Access  | Roundtable: Efficient and ethical genome data sharing Manuel Corpas, Scientific Lead, Repositive Fiona Nielson, CEO, DNA Digest, CEO, Repositive   | Roundtable: Defeating aging through AI Alex Zhavoronkov, (CSO, The Biogerontology Research Foundation) CEO, InSilico Medicine Inc   |
|-------|---|--|---|
| 12:30 |   | Networking Lunch   |   |
| 13:50 | Uniting the translational and clinical research communities by developing an integrated i2b2/tranSMART platform in the cloud  Sharing clinical data on a grand scale How to create a unilateral data sharing and collaborative system The future of TranSMART Keith Elliston, Chief Executive Officer, TranSMART Foundation   | How to drive NGS into the clinic  What are the roadblocks to driving NGS into the clinic?  How systems are allowing for the provision of a personalized healthcare system The greater picture  Wendy Rubinstein, Division Director, Clinical Data Management and Curation, ASCO's CancerLinQ Formerly, Chief, Medical Genetics and Human Variation, NCBI and Director, NIH Genetic Testing Registry (GTR)  | Artificial Intelligence in Drug Discovery and Aging Research  Insilicos Next generation mechanisms for drug development using Artificial Intelligence to discover new targets.  Breaking innovation stagnation in pharmaceuticals with Al  Working beyond human cognition and innovation using Insilco mechanisms  Alex Zhavoronkov, (CSO, The Biogerontology Research Foundation) CEO, InSilico Medicine Inc |
| 14:10 | Genomics: Improving Scientific Insights from Clinical Trials  • Advances in genome sequencing technologies have driven a dramatic increase in collecting genomics data, where genome sequencing in clinical trials is one of the fast growing applications.  • Industry-wide pain points are slowing the streamlined integration of genomics data into clinical trials.  • Clinical Trial Genomics provides at-scale, secure upload of genomics data and automated linking with study clinical data, machine-learning standardization of both data across studies, and turnkey analytics for immediately actionable hypotheses for on-going studies.  Jason Mezey, PhD, Senior Computational Biologist Contractor, Medidata Solutions | Oncology RWE, Hope, Dreams and Hard Realities  Big data has the potential to drive powerful insights in Cancer Care Clinical trials enroll only 3% of cancer patients, we need to learn from every patient Oncologists are seeking a "rapid learning system" that can democratize access to the most current clinical information.  Kevin Fitzpatrick, CEO, Cancer LinQ  An Innovative Approach to Improve | Defeating aging through genomics  • Effective antiaging methodologies through big data and genomics  • Understanding systems using genome wide association studies to allude to new mechanisms to aging.  |

### RNA-therapeutics bring RNA-seq into focus

- RNA therapeutics are innovative drugs to modulate the splicing and stability of specific RNA sequences
- Envisagenics
   SpliceCore<sup>™</sup> is a
   cloud-based platform
   for the discovery of
   druggable splicing
   events
- SpliceCore combines RNA-seq analysis with public data and machine learning to predict disease-causing splicing events and their regulators

Martin Akerman, CTO, Scientific Collaborator at CSHL, Envisagenics and Cold Spring Harbor Laboratory

# Cancer Care Through Evidence-Based Technology: NCCN and FlatIron collaborate on NCCN Quality & Outcomes Database

- Proving oncology stakeholders, the ability to garner critical insights needed to make informed decisions
- Electronic health record (EHR) data aggregated for cancer quality and outcomes assessment
- Leveraging cancer data in a meaningful way to identify opportunities to enhance and improve care

Elizabeth Nardi, Quality of Oncology Care Fellow, National Comprehensive Cancer Network

&

**Ken Carson,** Senior Medical Director, **Flatiron Health** 

- Where do we need the most help?
- Heart disease, cancer, diabetes
- Where technology comes in
- Cost of technology is exponentially decreasing, ushering in a new era in healthcare
- Looking at current treatment methods and where tech comes into play for heart disease, cancer, and type II diabetes. With special focus on:
- Heart disease: machine learning putting data to
- Cancer: early detection through deep learning
- Type II diabetes: digital therapeutics
- Creating big markets
- Rethinking clinical tests, from population based to time based
- Telemedicine
- On demand medicine
- Claims and interoperability
- Implications
- Software is ushering in a new era of prevention: (early) diagnostics, (wearable) instruments, (digital) therapies
- ♦ 10-year outlook

**Vijay Pande,** Professor, **Stanford** General Partner, **Andreessen Horowitz** 

## 14:50 New paradigms to enhance breast health through big data and genomics

- Using Big Data and Genomics to understand Germline risks developing breast cancer and triage.
- Breast cancer diagnosis and disease genomics for treatment and prognosis

Laura van 't Veer, Professor

#### Panel by [[LOGO]] ZS Associates

**Panel Discussion:** Big Data is taking the guesswork out of medicine

- How do you identify where to apply Big Data analytics in a Pharma company?
- How can teams accelerate new insights and tap into data that is there, but not accessible today?
- Can Big Data really change the way scientists, engineers and

PANEL DISCUSSION: Horizon Scoping the future of AI in pharmaceuticals and healthcare

**Jeff Evelhoch,** Vice President, Head of Translational Biomarkers, **Merck** 

**Michael Elashoff,** VP of Data Science, **MEDIDATA Solutions** 

Janusz Dutkowski, CEO, Data4Cure

|                | Laboratory Medicine and Associate Director Applied Genomics for the Cancer Center  | commercial teams support<br>drug development?  • How is running a big data  | Jennifer Campbell, Analyst, Union Square Ventures   |
|----------------|--|---|---|
|                | at <b>UCSF</b> ; co-founder <b>Agendia</b>   | project different from legacy<br>data warehouses and  |   |
| 15:10          | High Frequency Genomics:   | business intelligence?  |   |
|                | Direct-To-Patient studies  | Chair: <b>Mahmood Majeed,</b> Managing  |   |
|                | power advances in  | Partner Global Business   |   |
|                | monitoring disease activity &  | Technology/Transformation   |   |
|                | therapy response   | Executive/Data and Analytics Strategy   |   |
|                | transcriptomics  | Partner, <b>ZS Associates</b>   |   |
|                | The value of disease   |   |   |
|                | activity monitoring and  | Scott Bean, Director of Enterprise  |   |
|                | therapy monitoring<br>using blood-based  | Business Intelligence, Amgen  |   |
|                | transcriptomics  | Coursi Dai Diversaria e funda manatia e   |   |
|                | Challenges to making   | Suraj Pai Director of Information Systems, Amgen  |   |
|                | immune   | Systems, Amgen  |   |
|                | transcriptomics feasible   | Sandeep Varma, Principal, ZS  |   |
|                | <ul> <li>Using Direct-to-Patient</li> </ul>  | Associates  |   |
|                | sampling to drive larger   |   |   |
|                | cohorts of subjects  | Nagesh Bhide, Associate Director of   |   |
|                | Key technologies to  | Commercial Operations, Intercept  |   |
|                | enable High-Frequency<br>Transcriptomics   | Pharmaceuticals   |   |
|                | Transcriptomics  |   |   |
|                | <b>Bob Terbrueggen,</b> Founder &  |   |   |
|                | CEO, Dxterity Diagnostics Inc  |   |   |
|                |  |   |   |
| 15:30          | Afternoon refreshments   |   |   |
| 15:30<br>15:50 | Afternoon refreshments  FDA preparedness for "next gen   | Enabling Pediatric Precision Genomics   | Data Blitz  |
|                | FDA preparedness for "next gen sequencing"   | Rare disease research in  | Showcasing a selection of AI for  |
|                | FDA preparedness for "next gen sequencing"  • IT/Bioinformatics tools  | <ul> <li>Rare disease research in pediatrics requires</li> </ul>  |   |
|                | FDA preparedness for "next gen sequencing"  • IT/Bioinformatics tools developed at FDA to  | <ul> <li>Rare disease research in<br/>pediatrics requires<br/>collaborative networks</li> </ul>   | Showcasing a selection of AI for specific targets   |
|                | FDA preparedness for "next gen sequencing"  IT/Bioinformatics tools developed at FDA to support research and   | <ul> <li>Rare disease research in pediatrics requires collaborative networks</li> <li>Networks need to enable</li> </ul>  | Showcasing a selection of AI for specific targets  15:50 Numerate   |
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|                | FDA preparedness for "next gen sequencing"  IT/Bioinformatics tools developed at FDA to support research and regulatory needs  FDA research supporting regulatory  | <ul> <li>Rare disease research in pediatrics requires collaborative networks</li> <li>Networks need to enable institutions as well as investigators</li> <li>Academic institutions benefit</li> </ul>   | Showcasing a selection of AI for specific targets  15:50 Numerate Rethinking predictive modeling in the age of AI  • We need to change the way pharma thinks about AI. It needs to take more  |
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|       | geno-pheno analysis<br>and faster time to   | Governance, Sage Bionetworks   | attrition (ADME and tox).  • Given these successes,  |
|-------|---|--|--|
|       | actionable insights   |  | the models are still<br>fundamentally data<br>starved. We need to  |
|       | George Asimenos, PhD Chief Technology Officer, DNAnexus   |  | started. We need to  |
| 16:30 | Integrative analysis, including genome-wide expression data, in Neuroblastoma   | A perspective for NGS based cancer diagnostics; assay development, validation and compliance in the  | Brandon Allgood, CTO and a cofounder, Numerate Inc   |
|       | Integrative analysis of expression data with genomic  | midst of current and the future of Genomics  | 16:05 Enlitic  |
|       | information or with high-throughput drug screening  | Clinical trials and research     while focusing on enhancing   | Kevin Lyman, Chief Data Scientist,<br>Enlitic  |
|       | data provides for:  • Linking genomic  changes driving cancer   | <ul><li>cancer care delivery</li><li>Improving cancer outcomes</li><li>through a targeted treatment</li></ul>  | 16:20 Lunit  |
|       | to their consequences for biological processes  Testing predictive  | <ul><li>approach</li><li>How we respond to findings<br/>through personalized,</li></ul>  | <b>Brandon Suh,</b> Chief Medical Officer, <b>Lunit</b>  |
|       | biomarkers based on gene expression   | cancer-specific treatment plans.   | 16:35 Atomwise   |
|       | <ul> <li>Development of<br/>predictive biomarkers<br/>using machine learning</li> </ul>   | Pravin Mishra, Director, Precision Genomics Core Laboratory & R&D, Intermountain Healthcare, Dixie   | Abraham Heifets, CEO, Atomwise   |
|       | Jeffrey Bond, Lead<br>Bioinformatician, NMTRC,  | Regional Medical Center  |  |
| 16:50 | Spectrum Health System  How the cloud is affecting big  |  | Supercomputing and the Future  |
|       | data sharing?  Storing communicating and tracking your personal health records  Patient privacy and mechanisms  Data value and management with the patient at the center  Andreas M. Kogelnik, Director, CEO, Open Medicine Institute | The Joint Analysis of Many Matrices via ITeration (JAMMIT): Tailoring precise treatment strategies for cancer  • High-dimensional data sets of measurements for different data types obtained from a common set of biosamples are proliferating at an exponential rate in public and private databases • The complexity of such "multi-modal" data has slowed the development of new predictive biomarkers and treatments for cancer • Sparse, rank-1 matrix approximations implemented by the JAMMIT algorithm provide a simple and powerful approach to extracting predictive signatures from multi-modal data • JAMMIT analysis of real experimental data for ovarian and liver cancer reveal connections between | of Health  The use of AI in drug development  How BERG uses artificial intelligence to analyze tissue samples and clinical data to model and understand diseases and guide drug discovery  Understanding why AI is an overdue disruption to drive innovation and pharmaceutical development  Slava Akmaev, Ph.D., Senior Vice President & Chief Analytics  Officer, BERG |

|       |  | sparse signatures, immune checkpoint signaling, and overall survival  Gordon Okimoto, Co-Director, University of Hawaii Cancer Center   |  |
|-------|--|---|--|
| 17:10 | Rare disease patient community engagement, the key to accelerate research to clinical impact, a case study  • Focused generation and aggregation of rare disease "big data" can close the gap of time between research and clinical impact  • empowering patient family communities as equal stakeholders can drive international collaboration and data sharing  • leveraging genetics/functional genomics of one rare disease can be leveraged to rare and common diseases of similar biology  Christina Waters, CEO, RARE | <ul> <li>Genomic literacy: Developing a minimalist curriculum for healthcare providers, i.e. teach me just what I need to know about genomics to practice precision medicine</li> <li>Skills: Incorporating practical, hands on experiences ordering tests, interpreting reports, communicating with patients</li> <li>Awareness: Providing opportunities to stay apprised of the latest applications of genomics in healthcare</li> <li>Jeanette McCarthy, Adjunct Associate Professor, Duke University</li> </ul> | Connecting tumor genomics with therapeutics through multi-dimensional network modules  • Cancer cell lines can model therapeutic responses, but only partially reflect tumor biology.  • Using MAGNETIC, a new method to integrate molecular profiling data using functional networks, we identify 219 gene modules in TCGA breast cancers that capture recurrent alterations, reveal new roles for H3K27 tri-methylation and accurately quantitate various cell types within the tumor microenvironment.  • We show that a significant portion of gene expression and methylation in tumors is poorly reproduced in cell lines due to differences in biology and microenvironment and MAGNETIC identifies therapeutic biomarkers that are robust to these differences. This work addresses a fundamental challenge in pharmacogenomics that can only be overcome by the joint analysis of patient and cell line data  Sourav Bandyopadhyay, Assistant Professor, UCSF |
| 19:00 | End of Conference Day One  |   |  |

| 08:45 | Producers opening remarks   |   |                                       |
|-------|---|---|---------------------------------------|
| 00.43 | Edward Glanville, Project Director  | r. Terrapinn  |                                       |
| 08:50 | Chair's Opening Remarks   | ,   |                                       |
| 00.50 |   | al World Evidence, Epidemiology <b>, Nov</b>                              | artis Pharmaceuticals                 |
| 09:00 | Athula Herath, Global Head of Real World Evidence, Epidemiology, Novartis Pharmaceuticals  Deriving population centric phenotypes using Big Data for synthesizing Real World Evidence |   |                                       |
|       | From the outset Medicin   | e had been a data science. Careful and synthesis of evidence to formulate | d intricate observation of individual |
|       | • • •   | patients they treat have been pioneer ways been at the core of Medicine.  | ed by medical practitioners and       |
|       | •   | e you through a journey to create a co                                    | oordinate system (statistical         |
|       | •   | alth outcomes) that facilitates objectiv                                  |                                       |
|       | precision on population   | scale Real World Evidence Data.   |                                       |
|       | Athula Herath, Global Head of Re  | al World Evidence, Epidemiology <b>, Nov</b>                              | artis Pharmaceuticals                 |
| 09:20 | Genomics in space   |   |                                       |
|       |   | An Open Access Multi-Omics database                                       | of living samples flown in space:     |
|       | from microbes to plants,  |   |                                       |
|       | •   | nformatics: Strategies to democratize                                     | omics data to reach the science       |
|       | community and public at   | <u> </u>  | flife edentation in seco              |
|       | When users become bot   | :: The emergence of new knowledge o                                       | filte adaptation in space             |
|       | Sylvain Costes, Lead scientist for 0  |   |                                       |
| 09:40 |   | s: making large-scale data usable   |                                       |
|       |   | oad is an untenable model in the large                                    | =                                     |
|       | •   | ises that do not update the alignment                                     | and/or calling as technology          |
|       | advances quickly become   |   |                                       |
|       |   | Commons tries to solve those issues by                                    |                                       |
|       | database that will allow users to do queries online without the need to download the bulk of the  |   |                                       |
|       | raw data.  Jean Zenklusen, Director, The Cancer Genome Atlas, Centre for Cancer Genomics, Office of the Director,   |   |                                       |
|       | National Cancer Institute   |   |                                       |
| 10:00 | Morning refreshments  | Ī   |                                       |
|       | GENOMIC   | PRECISION   | ARTIFICIAL                            |
|       | S AND   | MEDICINE  | INTELLIGENCE                          |
|       | HEALTH  |   |                                       |
|       | Day 2   | Day 2 Chair:  | Day 2 Chair:                          |
|       | Day 2   | Jennifer Hall, Chief,   | Chair: Alex                           |
|       | Chair:  | Institute of Precision  | Zhavoronkov,                          |
|       | Chair: Manuel   | Cardiovascular Medicine,  | (CSO, The                             |
|       | Corpas, Scientific  | American Heart  | Biogerontolog                         |
|       | Lead, Repositive  | Association   | y Research                            |
|       | Lead, Nepositive  |   | Foundation)                           |
|       |   |   | CEO, InSilico                         |
|       |   |   | Medicine Inc                          |
|       |   | 1   | I IVICUICITE IIIC                     |

10:40 **PANEL DISCUSSION: Data** PANEL DISCUSSION: How to drive Deep learning in medicine: an **Democratization: Empowering** Precision medicine into the clinic introduction and applications to efficient access and sharing of Chaired by: Bill Barnett, CRIO, next-generation sequencing and Regenstrief and IU School of disease diagnostics Medicine, Regenstrief, Inc Andreas M. Kogelnik, Director, We review the history and **CEO, Open Medicine Institute** Laura van 't Veer, Professor taxonomy of machine Noel Burtt. Associate Director learning and artificial Laboratory Medicine and Associate of Operations, Broad Institute Director Applied Genomics for the intelligence Cancer Center, UCSF; co-founder, We will introduce deep Agendi learning, covering both Marie-Pierre Dubé, Director, what it is and why its so Beaulieu-Saucier Université de exciting. Montréal Pharmacogenomics We will then discuss in Centre detail two concrete Catherine Brownstein, Scientific applications to life sciences Director, Manton Center for Orphan problems: Calling SNP and indel Disease Research **Bob Terbrueggen,** Founder & variants in next-generation sequencing data CEO, Dxterity Diagnostics Inc Detection of diabetic retinopathy from fundus 11:20 Understanding the non-coding Big Data, Genomics and images of the eye genome Personalised Medicine: Future How much new paths for the AHA Jeff Dean, Senior Fellow, Google information do we **AHA launches MY** expect from the RESEARCH LEGACY - AN non-coding human **OPPORTUNITY FOR ALL** genome INDIVIDUALS TO ENGAGE IN Why is exome LIFELONG LEARNING TO sequencing not **IMPROVE THEIR OWN** enough? HEALTH AND THE HEALTH Where are the OF THOSE AROUND THEM, pathogenic variants in through biosensors, the non-coding technology and community. genome? "the Individual" Amalio Telenti, MD, PhD, CSO, AHA strategic partners and **Human Longevity** the "tech platform" underneath My Research Legacy The data from My Research Legacy - driving towards solutions for millions of patients Jennifer Hall, Chief, Institute of

Precision Cardiovascular Medicine,

American Heart Association

| 11.40 | Using big data and AI to drive   |
|-------|--|
|       | clinical drug development:<br>deep subtyping of disease;<br>continuous knowledge |
|       | deep subtyping of disease;   |
|       | continuous knowledge   |
|       | integration  |
|       |  |

- Integrating multimodal genomic and clinical data (public and proprietary) to build dynamic data-driven maps for hundreds of disease conditions
- Deep molecular and immune-infiltrate characterization of disease subtypes based on thousands of patient samples
- Biologically-guided machine learning to predict response to single agents and drug combinations

Janusz Dutkowski PhD, CEO, Data4Cure

#### Emerging Opportunities for Genomic Big Data Analytics in the Plecosystem Economy

- Critical opportunities for blockchain in Cancer Genomics: Micro-credit Accounting for data donors, institutional data users, ontology annotators, somatic read annotators, decision support algorithms etc.
- Emerging methods for early detection of onset and recurrence: The specificity/sensitivity dilemma
- Critical need for biomarkers across the plecosystem that help disambiguate isolated genomic data.

John Mattison, Assistant Medical Director/CMIO, Kaiser Permanente

## Building the Genomic Infrastructure that Powers Personalized Medicine

- Utilizing FPGAs in the cloud for highly efficient, optimized and low-cost data analysis
- Creating a true sample to answer platform
- Aggregating omics data for machine learning and Al

Emerging methods for early detection of onset and detection of onset



#### 12:00

#### **NETWORKING LUNCH**

## 13:20 Realizing the transformative potential of genomics in healthcare

Multimodality
 genomics for
 translational purposes
 Graphical molecular
 network analysis for

identification of novel

disease biology
 Diagnosis and clinical care of patients with inherited cardiovascular disease

Rick Dewey, Senior Director, Head of Translational Genetics, Regeneron Genetics Center

# The Big data hype: A national strategy for next generation genome health research, case Finland

- Is big data smoke and mirrors or does it really have a place in modern healthcare
- Breaking from the conscious and understanding data sets that are beyond comprehension without big data.
- Seeing the big data picture and treatment of diseases that have not been understood until the big data revolution in healthcare.

Aarno Palotie, Research Director, Institute for Molecular Medicine Finland (FIMM), Broad Institute of MIT and Harvard, Massachusetts General Hospital

# Big data on a network: Massive integration of domain knowledge to inform drug repurposing

- Resources spent on drug development are exorbitant. In parallel, the probabilities of a lead compound making it to clinic are minuscule.
- Developing a framework to integrate millions of experimental and clinical results in the form of a heterogeneous network, in which drugs, diseases, genes, etc are connected by mining a vast space of the entire domain knowledge.
- Using Machine learning to compute the probability that any given drug would interfere with mechanisms of a disease of interest (as a proxy for a potential therapeutic).

| 13:40 | 10 Simple Rules for Sharing Human Genome Data These 10 Simple Rules have been developed from our combined experiences with the Repositive platform, working with human genomic data, data repositories and data users. We do not claim that these rules will eliminate every possible risk of data misuse. Rather, we hope that these will help scientists to increase the reusability of their human genomic data, whilst also ensuring that the privacy of their subjects is maintained according to their consent frameworks. Many of the principles presented are also applicable to other types of clinical research data, where participant privacy is a concern.  Manuel Corpas, Scientific Lead, Repositive | Advancing discoveries in cardiovascular precision medicine through big data  AHA launches the Precision Medicine Platform Partnership with Amazon Web Services New approach to open data and tools and turn the attention to a community effort to accelerate solutions that positively impact the lives of those with cardiovascular disease and stroke.  Prad Prasoon, Business Technology Strategist at American Heart Association   | Sergio E Baranzini, Professor Weill Institute for Neurosciences, UCSF  "Leveraging Wearables in Clinical Trials"  • Learn how sensors and wearables are helping Pharmaceutical organizations foster creative, quality clinical trials using sensors and other wearable devices. • Learn how mobile devices can assist in the tracking and reporting of accurate clinical data • Discuss how technologies are now improving the clinical trial process and reduce costs and improve accuracy of clinical data in an expeditious manner  Bob Rogers PHD Chief Healthcare Data Scientist, Intel Corp |
|-------|---|---|---|
| 14:00 | Big data in the clinic – entering a new legal environment  Discussing the rapidly evolving legal and regulatory environment big data will encounter as it is integrated into clinical practice  Describing the potential legal landmines, and techniques to avoid them  Introducing risk management practices to minimize risk and maximize value  James Mills Barbeau, Assoc.  Professor, Brown University  Alpert Medical School  Director of Laboratory  Medicine,  Lifespan Academic Medical  | Novel sequencing-based assays as biomarkers of disease  Prediction of novel biomarkers using big data Genomic technologies to identify the genetic etiology and underlying mechanisms of human disease in order to define precision therapies for diseased individuals Predictive genomic signatures of response to therapy, and novel sequencing-based assays as biomarkers of disease Ali Torkamani, Director of Genome Informatics and Drug Discovery, The Scripps Translational Science Institute | When small data = big data, or the magic of transfer learning.  Sharing and connecting deep learning algorithms algorithms to create conditions for a cross-fertilization between powerful artificial intelligence systems?  Transfer learning to foster collaborative AI  How to bring big-data-trained deep learning algorithms into the world of medical data  How collaborative AI can bring new business models to create value with data  Gilles Wainrib, co-founder,   |

**Lifespan Academic Medical** 

Gilles Wainrib, co-founder,

|                | Center  |   | OWKIN  |
|----------------|---|---|--|
| 14:20          | Design and implementation of healthcare enterprise big data platforms  Developing and managing a comprehensive enterprise data strategy  Modern data governance strategies  Understanding the HPC landscape in healthcare and pharmaceutical development  Nicholas Marko, Founding board member of the International Society for Chief Data Officers, Director of Neurosurgical Oncology, Geisinger Health System | Metagenomic next-generation sequencing for pathogen detection  • Unbiased detection of pathogen nucleic acid from patient samples can be achieved through metagenomic next-generation sequencing (mNGS).  • Broad-based organism detection requires new approaches to validation and results interpretation.  • This talk will discuss the precision diagnosis of infectious disease for meningitis/encephalitis using mNGS.  Steve Miller, Director, UCSF  | Blockchain- a platform for Pharma The presentation will focus on where this potentially transformative platform for Pharmaceutical R&D could make the most direct benefitto patients  • How complexity could be simplified • Enabling trust • Allowing scale and security  Verner De Biasi, Head Emerging Platforms, GSK   |
|                |   | Clinical Microbiology Laboratory  |  |
| 14:40<br>15:20 | Afternoon refreshments Developing Inova's IT  | Discovering drivers of immune   | Al assistance for data science via   |
|                | infrastructure to support the collection, storage, visualization and distribution of genomic, clinical and laboratory data  • How to get the most value of large datasets quickly. • Key infrastructure tools and lessons learned. • Talent and resources needed to support research data services and analytics  Aaron Black, Chief Data Officer, Inova Translational Medicine Institute                         | response to cancer discovered through 'big data' analysis  Mutations in cancer not only drive the growth of tumors (driver mutations), but also help tumors to control host immune system (immunity driver mutations)  TCGA data analysis yields over 100 genetic regions that affect the immune response to cancer.  This discovery opens new direction for cancer immunology research and understanding individual responses to cancer immunotherapy  Adam Godzik, Director,  Sanford-Burnham Medical  Research Institute | <ul> <li>probabilistic programming</li> <li>Demand for data science is rapidly growing. However, many commercial and scientific data sources present fundamental inferential challenges.</li> <li>This talk will describe BayesDB, a probabilistic programming platform for Al-assisted data science that has been developed over the last 10 years.</li> <li>Novice BayesDB users can answer data analysis questions in seconds or minutes with a level of rigor that otherwise requires hours or days of work by someone with advanced training in statistics plus good statistical judgment.</li> <li>BayesDB also provides advanced probabilistic programming capabilities that enable experts to integrate</li> </ul> |

causal domain knowledge and

| 15:50 | Combining genomic, structural, and clinical data to discover new insights in hypertrophic cardiomyopathy  • We develop a new method to integrate structural features into tests of disease burden, especially in rare inherited diseases. • We use data from over 100,000 exomes and 2,900 rare disease patients to find structural regions of cardiac myosin enriched for mutations in disease patients. • We identify domains and surfaces of the myosin gene (MYH7) that are associated with clinical phenotypes and outcomes. • We demonstrate that combining data from different fields can identify novel correlations | Driving next generation diagnostics and precision medicine into the clinic  | black-box machine learning with hierarchical Bayes.  Examples will be drawn from collaborations with philanthropic organizations such as the Bill & Melinda Gates Foundation.  Vikash Mansinghka, Principal Investigator, The MIT Probabilistic Computing Project  Deep Learning for Identification of Drug Targets in the Pharmacoepigenome of the Human CNS  Deep learning applications in pharmacogenomics  Computational methods for accurate prediction of CNS pharmacodynamic networks  Mapping human CNS pharmacodynamic networks in space and time based on the 4D Nucleome Project (NIH)  Gerald Higgins, Research Professor of Computational Medicine and Bioinformatics, University of Michigan Medical School |
|-------|--|---|---|
|       | Julian Homburger, Data<br>Scientist, Stanford.   |   |   |
| 16:10 | IT infrastructure to speed the delivery of Precision medicine  | Genomics and cardiovascular clinical trials   | Predictive analytics for mortality risk estimation  |
|       | <ul> <li>into the clinic</li> <li>Developing an efficient network architecture to empower research</li> <li>New requirements for the big data era</li> <li>Customized working environments</li> <li>Jim Broach, Director of the Penn State Institute for</li> </ul>  | <ul> <li>Genomics for cardiovascular drug discovery</li> <li>Genomic studies of clinical trials</li> <li>Precision medicine of the CETP-inhibitor dalcetrapib</li> <li>Marie-Pierre Dubé, Director, Beaulieu-Saucier Université de</li> </ul> | <ul> <li>Machine learning techniques for survival analysis</li> <li>Joint modeling of longitudinal risk factors and survival data</li> <li>Biomarker-based mortality risk scores</li> <li>Statistical methodologies for</li> </ul>  |

|               | Personalized Medicine, University of Pennsylvania School of Medicine  | Montréal Pharmacogenomics<br>Centre   | studying longitudinal<br>dynamics of aging<br><b>Ghalib Bello,</b> Assistant  |
|---------------|---|---|---|
| 16:30         | Genomics and Health: Transferring the power of HPC  & NGS to the clinic  • Moving research to the clinic when working with mental health disorders  • The delicate balance of data sharing, discovery, and translation to the clinic  • Closing the loop- the transition back to focusing on the patient.  • Case studies of big data projects at a children's hospital  Catherine Brownstein, Scientific Director, Manton Center for Orphan Disease Research | Finding a needle in a haystack: new approaches to discover disease-causing mutations in patients' genomes  Prioritizing disease-causing candidate genes and mutations.  Detecting genotypic heterogeneity underlying phenotypic homogeneity. Filtering out Next Generation Sequencing false positives noise. Yuval Itan, Research Assistant Professor, Rockefeller University | Professor,Mount Sinai  Automated Genome-Based Prediction tool for Pathogens for the prediction of complex virulence and antibiotic resistance phenotypes using high throughput sequencing data  • Detection of antibiotic resistance phenotypes using high throughput sequencing data. • Machine learning algorithms to determine the diverse features (change in virulence genes, recombination, horizontal gene transfer, patient diagnostics). • Pathogenic Potential and Countermeasures Targets  Debjit Ray, Postdoctoral researcher, Sandia National Labs |
| 16:50         | Cannabis Strains and regulation  Understanding the current issues with regulation in cannabis  Producing and monitoring cannabis using databases and block chain  Big Data and Cannabis Kevin McKernan, CSO, Courtagen Life Sciences Inc  |   | Young Talent in Al 2017 Award and Presentation (showcasing the best young female talent in Al)  Duygu Tosun-Turgut, Assistant Professor and Co-Director of the Center for Imaging of Neurodegenerative Diseases (CIND), San Francisco Veterans Affairs Medical Center   |
| Save The Date |   |   |   |

Save The Date

### See you on the 13-14th March 2018 in San Francisco

Close of conference 17:10