



**Global Genomic  
Medicine Collaborative**

**3<sup>rd</sup> GLOBAL GENOMIC MEDICINE  
COLLABORATIVE (G2MC) CONFERENCE**



**IMPLEMENTING GENOMIC  
MEDICINE INTO PRACTICE**

**APRIL 27-29 2017**

**DIVANI APOLLON PALACE AND THALASSO, ATHENS, GREECE**



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Global Genomic  
Medicine Collaborative

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# Global Genomic Medicine Collaborative

## 3<sup>rd</sup> GLOBAL GENOMIC MEDICINE COLLABORATIVE CONFERENCE

### CONFERENCE ORGANIZING COMMITTEE

Geoff Ginsburg (Durham, NC, USA)  
Robyn Ward (Melbourne, Australia; Co-chair)  
George P. Patrinos (Patras, Greece; Local Chair)

Fahd Al-Mulla (Safat, Kuwait)  
Vajira Dissanayake (Colombo, Sri Lanka)  
Peter Goodhand (Montreal, Canada)  
Theodora Katsila (Patras, Greece)  
Bruce Korf (Birmingham, AL, USA)  
Teri Manolio (Baltimore, MD, USA)  
Alan Shuldiner (Tarrytown, NY, USA)

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### KEYNOTE SPEAKERS

Victor Dzau (Washington DC, USA)  
Stylianos Antonarakis (Geneva, Switzerland)

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### SPEAKERS AND MODERATORS

Takeya Adachi (Tokyo, Japan)  
Spiros Agathos (Urcuqui, Ecuador)  
Fahd Al-Mulla (Safat, Kuwait)  
François Bernier (Toronto, Canada)

Je\_rey Braithwaite (Melbourne, Australia)  
Mark Caul\_eld (London, UK)  
Wasun Chantratita (Bangkok, Thailand)  
Collet Dandara (Cape Town, South Africa)  
Vajira Dissanayake (Colombo, Sri Lanka)  
Geoff Ginsburg (Durham, NC, USA)  
Peter Goodhand (Montreal, Canada)  
Sue Hill (London, UK)  
Magnus Ingelman-Sundberg (Stockholm, Sweden)  
Federico Innocenti (Chapel Hill, NC, USA)  
Said Ismail (Doha, Qatar)  
Summer Kahlon (Satellite beach, FL, USA)  
Bruce Korf (Birmingham, AL, USA)  
Dhavendra Kumar (Cardiff, Wales)  
Guilherme Suarez-Kurtz (Rio de Janeiro, Brazil)  
Alberto Lecaros (Santiago, Chile)  
Yixue Li (Shanghai, P.R. China)  
Catalina Lopez-Correa (Quebec, Canada)  
Laura Lyman Rodriguez (Baltimore, MD, USA)  
Teri Manolio (Baltimore, MA, USA)  
George P. Patrinos (Patras, Greece)  
Markus Paulmichl (Vienna, Austria)  
Sir Munir Pirmohamed (Liverpool, UK)  
Martin Reese (Oakland, CA, USA)  
Gad Rennert (Haifa, Israel)  
Gabriella Repetto (Santiago, Chile)  
Ron H. van Schaik (Rotterdam, the Netherlands)  
Alan Shuldiner (Tarrytown, NY, USA)  
Andrew Sinclair (Melbourne, Australia)  
Jesse Swen (Leiden, the Netherlands)  
Patrick Tan (Durham, NC, USA)  
Domenica Taruscio (Rome, Italy)  
Aspasia Tsezou (Larissa, Greece)  
Joris Veltman (Newcastle, UK)  
Robyn Ward (Melbourne, Australia)  
Grant Wood (Montreal, Canada)  
Sarah Wordsworth (Oxford, UK)  
Marc S. Williams (Philadelphia, PA, USA)



# Global Genomic Medicine Collaborative

## 3<sup>rd</sup> GLOBAL GENOMIC MEDICINE COLLABORATIVE CONFERENCE

### SCIENTIFIC PROGRAM

[DIVANI Apollon Palace & Thalasso](#)

Leof. Vouliagmenis 10, Vouliagmeni, Athens 166 71 GREECE

DAY 1: APRIL 27, 2017

10:00–13:00

**Registration - [Location: Platon Foyer]**

13:00–13:10

**Welcoming Remarks - [Location: Platon]**

**Geoff Ginsburg**

*Director, Duke Center for Applied Genomics &  
Precision Medicine; Professor of Medicine, Biomedical  
Engineering and Pathology, Duke University, USA*

**Robyn Ward**

*Deputy Vice-Chancellor (Research) and Vice President  
(Research)*

The University of Queensland, Australia

**George P. Patrinos**

*Associate Professor of Pharmacogenomics and  
Pharmaceutical Biotechnology*

University of Patras, Greece

13:10-13:25

**Welcome from Hellenic Association of Medical  
Geneticists**

**[Location: Platon]**

**Aspasia Tsezou**

President

Hellenic Association of Medical Geneticists, Greece

13:25-13:55

**Keynote Lecture #1 [Location: Platon]**

**Victor Dzau**

President  
National Academy of Medicine, USA

**Session I: Large-Scale National Sequencing Programs: Impact on Clinical Medicine**

**Moderator: Alan Shuldiner, Vice President, Regeneron Genetics Center,  
Regeneron Pharmaceuticals, Inc., USA - [Location: Platon]**

- 14:00                    ***The Qatar Genome Program; An Overview***  
**Said Ismail**  
*Programme Manager*  
Genome Qatar, Qatar
- 14:15                    ***United States: PMI and Million Veterans Program***  
**Teri Manolio**  
*Director, Division of Genomic Medicine*  
National Human Genome Research Institute, USA
- 14:30                    ***China's Precision Medicine Initiative***  
**Yixue Li**  
*Professor*  
Shanghai Institutes for Biological Sciences, China
- 14:45                    ***UK: 100,000 Genomes Project***  
**Mark Caulfield**  
*Co-Director*  
William Harvey Research Institute  
Barts and the London School of Medicine and  
Dentistry, UK
- 15:00                    ***Australian Genomics Health Alliance:  
implementing genomics into health care***  
**Andrew Sinclair**  
*Deputy Director*  
Murdoch Children's Research Institute, Australia
- 15:15                    ***The Clalit Israeli 100K Personalized Medicine RCT***  
**Gad Rennert**  
*Director*  
Clalit National Israeli Cancer Control Center, Israel
- 15:30                    **Panel discussion**  
**Opening remarks: Chen-Yang Shen**  
*Chief Executive*  
Taiwan Biobank, Taiwan



16:00–16:15

**COFFEE BREAK**

**Session II: Progress in Diagnosing Rare Diseases – International Opportunities**

**Moderator: Teri Manolio, Director, Division of Genomic Medicine, National Human Genome Research Institute (USA) - [Location: Platon]**

16:15

**Undiagnosed severe intellectual disability**

**Joris Veltman**

*Director,*  
Institute of Genetic Medicine  
Newcastle University, UK

16:30

**Undiagnosed Diseases Network International**

**Domenica Taruscio**

*Director*  
Istituto Superiore di Sanita (ISS), Italy

16:45

**Next Generation Sequencing for rare pharmacogenomics variants**

**Magnus Ingelman-Sundberg**

*Professor*  
Karolinska Institutet, Sweden

17:00

**Japan's Initiative on Rare and Undiagnosed Diseases (IRUD): Towards integrative diagnosis network for universal healthcare system**

**Takeya Adachi**

*AMED Program Officer*  
Japan Agency for Medical Research & Development (AMED), Japan

17:15

**Enhanced Care for Rare Diseases Canada**

**François Bernier**

*Associate Professor,*  
Department of Medical Genetics  
Alberta Children's Hospital, Canada

17:30

**The University of Alabama at Birmingham Undiagnosed Diseases Program**

**Bruce Korf**

*Wayne H. and Sara Crews Finley Chair in Medical Genetics Professor and Chair, Department of Genetics*  
University of Alabama, Birmingham, USA

17:45 - 18:15

**Panel discussion**

18:30

**WELCOME COCKTAIL RECEPTION [Location: Swimming Pool]**

**DAY 2: APRIL 28, 2017**

08:15

**Opening Remarks - [Location: Platon]**

**Geoff Ginsburg**

*Director, Duke Center for Applied Genomics & Precision Medicine, Professor of Medicine, Biomedical Engineering and Pathology  
Duke University, USA*

**Robyn Ward**

*Deputy Vice-Chancellor (Research) and Vice President (Research)  
The University of Queensland, Australia*

08:20

**Keynote Introduction [Location: Platon]**

**George P. Patrinos**

*Associate Professor of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras, Greece*

08:25

**Keynote Lecture #2 [Location: Platon]**

**Stylianos Antonarakis**

*Professor and Chairman of Genetic Medicine  
University of Geneva Medical School, Switzerland*

**Session III: National Genomic Medicine Programs: Implementation**

**Moderator: Geoff Ginsburg, Director, Duke Center for Applied Genomics & Precision Medicine, Professor of Medicine, Biomedical Engineering and Pathology, Duke University - [Location: Platon]**

09:00

**Kuwait Genome Project**

**Fahd Al-Mulla**

*Director of Genomic Medicine Center and Professor of Molecular Pathology  
Kuwait University, Dasman Diabetes Institute,  
Kuwait*

- 09:15 ***Implementing Genomic Medicine in Sri Lanka***  
**[pre-taped video presentation]**  
**Vajira Dissanayake**  
*Chair & Professor of Anatomy and Medical Geneticist*  
University of Colombo, Sri Lanka
- 09:30 ***South America***  
**Gabriella Repetto**  
*Director, Center for Genetics and Genomics*  
Universidad del Desarrollo, Chile
- 09:45 ***The economics of next-generation sequencing***  
**Sarah Wordsworth**  
*Associate Professor,*  
Oxford University, UK
- 10:00 ***National Scale Precision Medicine in Singapore***  
**Patrick Tan**  
*Professor, Cancer and Stem Cell Biology Program*  
Duke NUS Medical School, Singapore
- 10:15 ***IGNITE network***  
**Geoff Ginsburg**  
*Director, Duke Center for Applied Genomics &*  
*Precision Medicine; Professor of Medicine,*  
*Biomedical Engineering and Pathology*  
Duke University, USA
- 10:30 **Panel discussion**

## Session IV: Data Science (with GA4GH)

**Moderators: Peter Goodhand, Executive Director, Global Alliance for Genomics and Health, Grant Wood, Senior IT Strategist, Clinical Genetics Institute at Intermountain Healthcare - [Location: Platon]**

- 11:00 ***Accurate and Rapid WGS Interpretation with Fabric Genomics***  
**Martin Reese**  
*Co-founder, President, and Chief Scientific Officer*  
Fabric Genomics, USA
- 11:15 ***Clinical contextualization of lab results***  
**Marc S. Williams**  
*Director, Genomic Medicine Institute*  
Geisinger Health System, USA

- 11:30 **Oracle Family Health History Project**  
**Summer Kahlon**  
*Director, Care Innovation*  
Oracle Health Sciences, USA  
**Grant Wood**  
*Senior Strategist*  
Intermountain Healthcare Clinical Genetics Institute,  
USA
- 11:45 **Establishing a Longitudinal Genetic/Genomic-  
Based EHR for Clinical Care**  
**Peter Goodhand**  
*President, Ontario Institute for Cancer Research*  
*Executive Director, Global Alliance for Genomics and  
Health, Canada*
- 12:00 **Q&A with session speakers and audience**  
Discussion topic: What are the issues in your country  
to accomplish the goals of a longitudinal genomic  
patient record?
- 12:30 **LUNCH BREAK - [Location: Platon foyer]**

## Session V: Policy and Workforce Generation

**Moderator: Robyn Ward, Deputy Vice-Chancellor (Research) and Vice President (Research), The University of Queensland, Australia, and Bruce Korf, Wayne H. and Sara Crews Finley Chair in Medical Genetics, Professor and Chair, Department of Genetics, University of Alabama, Birmingham [Location: Platon]**

- 13:30 **The Implementation of Genomic Medicine in the  
NHS in England**  
**Sue Hill**  
*Chief Scientific Officer*  
NHS England, UK
- 13:45 **Genomic Test Evaluation Frameworks**  
**Robyn Ward**  
*Deputy Vice-Chancellor of Research, and Vice  
President of Research*  
The University of Queensland, Australia

- 14:00 ***Implementing Genomics in the Health Care System in Canada***  
**Catalina Lopez-Correa**  
*Chief Scientific Officer and Vice President, Sectors Genome British Columbia, Canada*
- 14:15 ***More than regulatory challenges for genomic medicine in Latin America***  
**Alberto Lecaros**  
*Observatory for BioEthics and Law  
Universidad del Desarrollo  
Santiago, Chile*
- 14:30 ***Establishing a Latin American Translational Genomics Institute***  
**Spiros Agathos**  
*Dean, School of Life Sciences  
Yachay Tech University, Ecuador*
- 14:45 ***Implementation Science Meets Genomic Medicine: Ideas, Issues & Innovations***  
**Jeffrey Braithwaite**  
*Professor of Health Systems Research, Founding Director, Australian Institute of Health Innovation  
Macquarie University, Australia*
- 15:00 ***Fostering Genomic Literacy for Today and Tomorrow***  
**Laura Lyman Rodriguez**  
*Director, Division of Policy, Communications, and Education, National Human Genome Research Institute (NHGRI), USA*
- 15:15 ***Genomic education for empowering the Multi-Disciplinary Healthcare***  
**Dhavendra Kumar**  
*Visiting Professor, Genomic Policy Unit, Faculty of Life Sciences & Education, University of South Wales, UK; Consultant in Clinical Genetics, University Hospital of Wales, UK*
- 15:30 **Panel discussion**
- 16:00 **BREAK**

16:15–17:30

**Breakout Sessions**

**A) National Programs: Implementation -  
[Location: Platon]**

**Chair: Alan Shuldiner**

Vice President, Regeneron Genetics Center,  
Regeneron Pharmaceuticals, Inc., USA

**B) Pharmacogenomics - [Location: Platon]**

**Chair: George P. Patrinos**

Associate Professor of Pharmacogenomics and  
Pharmaceutical Biotechnology; University of  
Patras, Greece

**C) Policy [Location: Athina A]**

**Chair: Robyn Ward**

Deputy Vice-Chancellor of Research, and Vice  
President of Research, The University of  
Queensland, Australia

**D) Education and Workforce [Location: Athina  
B]**

**Chair: Bruce Korf**

Wayne H. and Sara Crews Finley Chair in Medical  
Genetics; Professor and Chair, Department of  
Genetics, University of Alabama, Birmingham,  
USA

**E) IT/Bioinformatics & Family Health History  
Project**

**[Location: Poseidon A]**

**Chairs: Peter Goodhand**

President, Ontario Institute for Cancer Research;  
Executive Director, Global Alliance for Genomics  
and Health, Canada

**Grant Wood**

Senior Strategist, Intermountain Healthcare Clinical  
Genetics Institute, USA

**F) Evidence [Location: Poseidon B]**

**Chairs: Marc Abramowicz**

Professor and Head, Dept. of Medical Genetics  
Université Libre de Bruxelles, Brussels

**Fahd Al-Mulla**

Director of Genomic Medicine Center and Professor  
of Molecular Pathology, Kuwait University,  
Dasman Diabetes Institute, Kuwait

18:00 **Depart for Dinner [meet in hotel lobby]**  
Tour of [Acropolis Museum](#)

20:00 **DINNER**  
Location: [Acropolis Museum](#)

### DAY 3: APRIL 29, 2017

07:00–08:00 **G2MC Steering Committee Meeting (by invitation only) - [Location: Poseidon A]**

08:00 **Opening Remarks - [Location: Platon]**  
**Geoff Ginsburg**  
Director, Duke Center for Applied Genomics & Precision Medicine; Professor of Medicine, Biomedical Engineering and Pathology  
Duke University, USA  
**Robyn Ward**  
Deputy Vice-Chancellor of Research, and Vice President of Research  
The University of Queensland, Australia

### Session VI: Pharmacogenomics in the Clinic: International Progress

**Moderator: George P. Patrinos, Assistant Professor of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras [Location: Platon]**

08:10 **Implementation of Pharmacogenomics in the Clinic**  
**Sir Munir Pirmohamed**  
*Professor*  
University of Liverpool, Wolfson Institute of Personalized, Medicine, UK

08:20 **Genomic Medicine, Safety First**  
**Federico Innocenti**  
*Associate Director, Center for Pharmacogenomics & Individualized Therapy, Associate Professor,*  
University of North Carolina Chapel Hill, USA

08:30 **The Ubiquitous Pharmacogenomics (UPGx) project**  
**Jesse Swen**  
Associate Professor, Department of Clinical Pharmacy & Toxicology  
Leiden University Medical Center, Netherlands

- 08:40 **PGx Activities in Latin America**  
**Guilherme Suarez-Kurtz**  
*Professor*  
Instituto Nacional de Câncer, Brazil
- 08:50 **Public Health Pharmacogenomics**  
**George P. Patrinos**  
*Associate Professor of Pharmacogenomics and  
Pharmaceutical Biotechnology*  
University of Patras, Greece
- 09:00 **Implementation of Pharmacogenomics in the  
Austrian Healthcare System**  
**Markus Paulmichl**  
*Medical Advisor*  
Centre for Health and Bioresources, Austrian  
Institute of Technology, Austria
- 09:10 **European Society of Pharmacogenomics**  
**Ron van Schaik**  
*Full Professor, Pharmacogenetics*  
Erasmus Medical Center, Netherlands
- 09:20 **Pharmacogenomics research and implementation  
in Africa**  
**Collet Dandara**  
*Professor, Department of Clinical & Laboratory  
Sciences, University of Cape Town, South Africa*
- 09:30 **The SEAPharm Consortium**  
**Wasun Chantratita**  
*Associate Professor, Department of Pathology  
Ramathibodi Hospital, Thailand*
- 09:40 **Panel discussion**
- 10:15-10:30 **BREAK**
- 10:30-12:00 **Breakout Reports and Discussion [Location:  
Platon]**  
**Moderator: Geoff Ginsburg**  
*Director, Duke Center for Applied Genomics &  
Precision Medicine; Professor of Medicine, Biomedical  
Engineering and Pathology*  
Duke University



12:00–13:00

**LUNCH [Location: Swimming Pool]**

**WORKING LUNCH [Location: Poseidon A]  
(G2MC Steering Committee Meeting - by invitation only)**

## Session VII: Implementation of Genomic Medicine by the Private Sector

**Moderator: Geoff Ginsburg, Director, Duke Center for Applied Genomics & Precision Medicine, Professor of Medicine, Biomedical Engineering and Pathology, Duke University [Location: Platon]**

13:15

***De novo assembly of Asian diploid genome (AK1)***

**Changhoon Kim**

*Director, Bioinformatics Institute  
Macrogen, South Korea*

13:30

***The Science May be the Easy Part: Challenges and Solutions in Implementing a Molecular Diagnostic Test***

**James Wingrove**

*Vice President, Technical Operations, Head of Research, CardioDx, USA*

13:45

***From human genetic insights to drug discovery impact***

**Nadeem Sarwar**

*President  
Eisai AiM Institute, USA*

14:00

***Genomic Medicine Enabled Through Partnership***

**David Bentley**

*Vice President and Chief Scientist  
Illumina, UK*

14:15

**Closing remarks**

**Geoff Ginsburg**

*Director, Duke Center for Applied Genomics & Precision Medicine, Professor of Medicine, Biomedical Engineering and Pathology, Duke University, USA*

**Robyn Ward**

*Deputy Vice-Chancellor (Research) and Vice President (Research)*

*The University of Queensland, Australia*

14:30

**MEETING ADJOURNS**





# Global Genomic Medicine Collaborative

## ABSTRACTS – INVITED SPEAKERS

### **Japan’s Initiative on Rare and Undiagnosed Diseases (IRUD): Towards integrative diagnosis network for universal healthcare system**

*Takeya Adachi*

AMED Program Officer, Japan Agency for Medical Research & Development (AMED)

To identify rare—and often undiagnosed—diseases, we must integrate systematic diagnosis by medical experts with patients’ phenotypic/genetic data matching, thereby solving the “N-of-1” problem. Initiative on Rare and Undiagnosed Diseases (IRUD), having been launched and coordinated by AMED since 2015, is an ambitious challenge in this context to construct a comprehensive medical network and establish useful clinical databases upon the Japanese universal healthcare system, resulting in 200 collaborating hospitals and 2200+ patients registered. We could successfully diagnosis 22% of patients prior to WES/WGS and identify the causative variants in 30-40% of the

rest by WES/WGS. Furthermore, Japan has already taken measures for specially defined rare diseases, called “Nan-Byo” in Japanese. IRUD is unique in that it takes advantage of this long-term asset. We believe in its significant contribution to the international endeavors, involving players in basic research, applied research, and social implementation.

### **Establishing a Latin American Translational Genomics Institute**

*Spiros N. Agathos and Juergen Reichardt*

Yachay Tech University, San Miguel de Urcuquí, Ecuador

Yachay Tech is the first research-intensive University in Ecuador, founded in 2014 as part of a national plan towards a knowledge-based economy. Within this initiative, Yachay Tech is establishing a Translational Genomics Institute in cooperation with the Golden Helix Foundation, a non-profit organization disseminating an understanding of human genetics and

promoting the clinical application of pharmacogenomics in medical practice. The two institutions will carry out research and teaching in Genomics related to Systems Biology, Bioinformatics, Population Genetics, Pharmacogenomics and Genomic & Translational Medicine. The 'Translational Genomics Institute' at Yachay Tech aspires to be a center of reference for Genomics in South America for analyzing genetic data on disease incidence and guiding research and public health policies of relevance to various populations and ethnic groups. In addition, the Institute will be a key partner in research projects addressing the tremendous wealth of the continent's biodiversity for novel biopharmaceuticals.

### **Genomic Medicine Enabled Through Partnership**

*David R Bentley & Collaborators*  
Illumina Inc.

Partnerships between medical, academic and commercial organisations are an essential feature of implementation of genomic medicine. In partnership with Genomics England and other collaborators, we have established an infrastructure for large scale whole genome sequencing (WGS) and its application to (a) rare and undiagnosed genetic disease and (b) cancer. To date ~30,000 genomes

have been returned as part of the 100,000 Genomes Project in the UK. Technology development is focused on sequencing clinical samples, improving variant calling, annotation and reporting, and in developing robust workflows for returning data quickly to the clinic. Utilization of PCR-free WGS data supports detection of complex germline variants (e.g. copy number variants and repeat expansions) and significantly increases diagnostic yield. WGS of cancer also maximizes detection of the range of somatic mutations (including structural rearrangements and mutational signatures) in cancer. The ecosystem for large scale WGS is applicable in national and regional situations to enable the effective implementation of WGS for genomic medicine.

### **Implementation Science Meets Genomic Medicine: Ideas, Issues & Innovations**

*Jeffrey Braithwaite*  
Australian Institute of Health Innovation, Macquarie University, Australia

The genomics revolution is well underway and the Global Genomic Medicine Collaborative (G2MC) has assembled an impressive array of collaborators in countries across the world to further the overarching objective of implementing genomic

medicine in clinical care. In this paper, the key word is “implementation” and the key phrase is “implementation science”. Implementation science is an embryonic discipline assembling theories, tools and translational ideas so that we can understand how take-up, adoption and spread of new practices work in different contexts and settings. In this presentation we discuss how implementation science is contributing to the G2MC’s work, and we look at a selection of models, studies and theories that can help take research from the bench-to-bedside. We are interested in providing tools that help us conceptualize improvement cycles over time, the spread of better practices and going full scale in adopting genomic medicine in clinical care.

### **Taiwan Biobank for the Health of the Next Generation**

*Chen-Yang Shen*

Taiwan BioBank, Academia Sinica, Taipei, Taiwan; Institute of Biomedical Sciences, Academia Sinica, Taipei, Taiwan

To understand the relationship between genetics, environmental exposure, diet, and the etiology/progression of chronic disease, the Taiwan Biobank (TWB) is establishing a scientific infrastructure accessible to biomedical researchers. Through the recruitment and follow-

up of a cohort of 200,000 individuals from the general population and another of 100,000 patients with chronic diseases from medical centers, the Taiwan Biobank aims to improve the health of future generations and facilitate genomic research in Taiwan. Currently, more than 80,000 participants from different parts of Taiwan have been recruited, and more than 1,750,000 of biospecimens, including blood, urine DNA and tumor tissues, had been collected. Whole-genome genotyping of more than 20,000 individuals using TWBv1.0 chip (653,291 SNPs, specifically for the Han Chinese in Taiwan) was obtained. An investigation of the population structure in an initial freeze of 10,801 unrelated TWB participants shows that the Taiwanese Han Chinese clustered into three cline groups: 5% were of northern Han Chinese ancestry, 79.9% were of southern Han Chinese ancestry, and 14.5% belonged to a third (T) group. We also find that this T group is genetically distinct from neighboring Southeast Asians and Austronesian tribes but similar to other southern Han Chinese. Interestingly, high degree of LD between HLA haplotype A\*33:03-B\*58:01 and SNPs across the MHC region was observed in subjects with T origin, but not in other Han Chinese. Furthermore, whole genome sequencing of 1,500 individuals was completed. This work aims at (1) establishing a local reference for imputation and (2) detecting

population-specific rare variants that would be of particular importance for the understanding of local diseases. Genomic features are currently under investigation. The release of TWB information and specimen would lead to the development of precision medicine in Taiwan, by which the progressive elucidation of risk factors and the molecular pathogenesis of disease will both improve disease prevention/prediction and facilitate therapy development.

### **Implementing Genomic Medicine in Sri Lanka**

*Vajira H. W. Dissanayake*

Medical Geneticist; Chair Professor of Anatomy; Director, Human Genetics Unit; Chair, Specialty Board in Biomedical Informatics, Faculty of Medicine, University of Colombo, Sri Lanka

Widespread adoption of Genomic Medicine in clinical practice would depend on effective deployment of next generation sequencing technologies and bioinformatics tools in relatively 'small' genetic centers. Recent advances in next generation sequencing technologies and the associated cost reduction in exome, clinical exome, and gene panel sequencing has made it possible for such centers to access these technologies. There are however many challenges that one would encounter

in the process of implementing Genomic Medicine. The Human Genetics Unit of the Faculty of Medicine, University of Colombo is one of the few centers in the South Asian region to have successfully implemented Genomic Medicine. We routinely sequence clinical exomes and cancer gene panels in house on Illumina's MiSeq platform and perform our own bioinformatics analysis on a bioinformatics pipeline established in house using free and open source tools. We have also established the protocol for providing genetic counseling using genomic test results. Although we have successfully implemented Genomic Medicine, in the process, we have had to experience challenges related to interpreting results (stemming from the fact that our population is not represented in international genetic databases), ethical issues, and issues related to cost and timeliness of accessing technical services (as they are delivered in countries like Sri Lanka through third parties). In this talk I shall share our experience with the participants. I hope that it would help understand what is required to take Genomic Medicine from large genome centers to small genetic clinics as well as inform what is required to ensure that there is equitable distribution of access to Genomic Medical services across the world to facilitate implementing Genomic Medicine globally.

## **The IGNITE network: a model for genomic medicine implementation and research**

Geoffrey S Ginsburg MD PhD  
Director, Duke Center for Applied Genomics and Precision Medicine  
Professor of Medicine, Pathology and Biomedical Engineering  
Duke University, USA

Challenges exist to widespread clinical implementation of genomic medicine, a prerequisite for developing evidence of its real-world utility. To address these challenges, the National Institutes of Health-funded IGNITE (Implementing GeNomics In pracTicE; [www.ignite-genomics.org](http://www.ignite-genomics.org)) Network, comprised of six projects and a coordinating center, was established in 2013 to support the development, investigation and dissemination of genomic medicine practice models that seamlessly integrate genomic data into the electronic health record and that deploy tools for point of care decision making. IGNITE site projects vary in scope and design, including exploring genetic markers for disease risk prediction and prevention, developing tools for using family history data, incorporating pharmacogenomic data into clinical care, refining disease diagnosis using sequence base mutation discovery, and creating novel educational approaches. Lessons learned and solutions to barriers in the implementation of genomic medicine programs will be described as well as

the development of a centralized 'tool box' for use by the genomic medicine community.

## **Implementation of rare genetic variants**

*Magnus Ingelman-Sundberg*  
Karolinska Institutet

Results from analyses of WGS and WES sequencing efforts reveal that about 40 % of all LOF and missense mutations of importance for prediction of drug response are rare. Pharmacokinetic analyses in twins also reveal that 50 % of the interindividual inherited variation in metoprolol and toresimide pharmacokinetics are caused by mutations not routinely analysed in the pharmacogenetic platforms currently used. The talk will consider the genes and drugs of importance where NGS based sequencing data would substantially add information beyond the current platforms to facilitate individualized drug therapy.

## **Genomic Medicine, Safety First**

*Federico Innocenti*  
Associate Director, Center for Pharmacogenomics and Individualized Therapy; Associate Professor, University of North Carolina Chapel Hill, USA

By putting the patient, rather than the disease, at the center of personalized care, the concept of safety gains a much wider connotation. Hippocrates' "primum non nocere", "first, do not harm" remains a fundamental guiding principle of any intervention in medical care. The suffering and discomfort experienced by patients because of the symptoms related to the adverse events of drug treatment should never be discounted. Adverse events have a significant burden on the quality of life of the patient and the families. They reduce confidence in the treatment and might demoralize the patient and the trust on the efficacy of therapies that should improve patient well-being, not reducing it. And we should not forget that, in this era of advanced technologies integrated into patient care, patients treated with anti-cancer therapies are still at risk of losing their lives as a result of the medication that should treat their cancers. Severe adverse events often lead to permanent discontinuation of therapy. Mild-to-moderate toxicities, in addition to affecting quality of life, can also reduce the intensity of the regimen. Most cancer patients receive multi-drug regimens, and toxicity from one drug can halt also the whole regimen. When the goal of treatment is to maintain acceptable quality of life and prolong survivorship (or survival), patient discomfort (from gastrointestinal effects) or cosmetic changes (acne or skin rash), for example, even when they are not

severe, might reduce adherence to oral therapy, increasing the chance of recurrence of disease. This presentation will describe the place for genetic analyses to increase patient safety, and how they have the potential of improving safety of cancer drugs in several ways. These aspects are of particular importance for implementation, as access to genetic profiling is becoming more common for patients.

### **The Qatar Genome Program; An Overview**

*Said Ismail*

Manager, Qatar Genome Program,  
Qatar Foundation

The Qatar Genome Program (QGP) is a national project spearheading the implementation of advanced precision medicine in Qatar. The project is designed around on a comprehensive strategy built on seven building blocks: the Qatar Biobank; building genomic infrastructure; drafting regulations and policies; forging networks of research partnerships; establishing national genomic data network; building local human capacity; and finally integration of genomics into the healthcare system. The pilot phase was launched in September 2015, with the goal of revealing the key features of the Qatari genome by sequencing 6,000 genomes by Mid 2017. Other pilot phase goals include promoting



genomic research in Qatar through research grants in addition to encouraging multicenter research teams to mine the data being produced. QGP is also taking the lead in building local human capacity through its internship programs, workshops and symposia, as well as taking the initiative in starting new graduate programs in genomic medicine and genetic counseling in collaboration with local universities. Additionally, QGP is involved in drafting a national policy on genomic research ethics and regulations.

### ***De novo assembly of Asian diploid genome (AK1)***

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With maturation of the recent high-throughput and/or single molecule sequencing technologies, the hope for medical usage of genome is ever increasing. Thus, as a part of Asian Genome Project, we built a new Asian reference genome AK1. In this work, we discuss the de novo construction of an Asian diploid genome with single molecule real time (SMRT) sequencing, single molecule optical-mapping, and microfluidic-based linked reads. The use of complementary single molecule technologies yields assembly with contig N50 of 17.95 Mb and scaffold N50 of 44.8 Mb. The AK1 assembly, notably, spans 7 chromosomal arms with essentially single scaffolds, showing high contiguity of the assembly and covers many gaps in the GRCh38. There are 18,210 structural variations (SVs), among which 5,294 insertions and 3,983 deletions were novel. Many of them were Asian specific SVs. In addition, the assembly was de novo phased by using the linked-reads and legacy BAC clones. The final phased block N50 was larger than 10 Mb. Our result shows ethnicity-specific, difficult-to-find variations can be effectively examined by constructing de novo assembly of diploid human genomes with long

range information and single molecule technologies.

### **The University of Alabama at Birmingham Undiagnosed Diseases Program**

*Bruce R. Korf*

Department of Genetics, University of Alabama at Birmingham, Birmingham, AL, USA

The UAB Undiagnosed Diseases Program (UDP) has been in operation since October 2013. It is not part of the NIH-funded Undiagnosed Diseases Network, and is funded directly by UAB. The program provides evaluations of both children and adults and includes four part-time physicians, two nurse-practitioners, a genetic counselor, and a genetic counseling assistant. There are also multiple consultants representing specialties in both pediatrics and adult medicine. Since its inception, the program has received 326 referrals. After review of records and triage of cases that the program does not deem appropriate for evaluation, 131 patients were enrolled for evaluation. At present 77 had completed evaluations, and diagnoses were achieved for 45. The program performs whole genome or whole exome sequencing on most patients, unless the nature of the clinical problem is such that this is deemed unproductive. Diagnoses have included genetic conditions,

unrecognized malignancy, and non-genetic medical conditions such as rheumatological or neurological diagnoses. Evaluations are performed on a fee-for-service basis, though genomic analysis is done on a research basis using institutional funds, and pathogenic findings are replicated in a CLIA and CAP certified clinical laboratory. UAB is now launching a new program, the “Alabama Genomic Health Initiative,” which is funded by the state of Alabama. This program will provide genotyping using the Illumina Global Screening Array on 10,000 individuals, with return of results of medically actionable pathogenic variants. In addition, we plan to offer genome sequencing to approximately 250 individuals with undiagnosed disorders as part of this program over the next five years. This will permit us to expand the genomic reach of the UDP, and also will result in a research database. In addition, we are planning to develop a drug screening program to focus on pathogenic variants identified in the UDP for which there are currently no therapeutic options.

### **Genomic education for empowering the Multi-Disciplinary Healthcare**

*Dhavendra Kumar*

Institute of Medical Genetics, University Hospital of Wales, Cardiff & The Genomic Policy Unit, The University of South Wales, UK

The treatment and management of rare genetic disorders require a coordinated and systematic effort of many skilled clinicians, nurses and other health professionals. The role of the clinical genetics team is now increasingly recognized in the multi-disciplinary team (MDT) approach in the treatment and management of rare chromosomal (for example The 22q Deletion Syndrome) and monogenic disorders (for example Mendelian Inherited Cardiovascular Conditions). Outcomes and experience of these model MDT approaches have been very positive with wide appreciation and acknowledgement across the UK National Health Service.

All MDT clinicians and health professionals provide invaluable input. Most MDT members have are encouraged and supported to acquire basic skills and competencies in eliciting and interpreting the family history and acquiring/assembling the necessary clinical and laboratory evidence for making the diagnosis and instituting the management plan.

Increasingly, all non-genetic MDT members are encouraged to learn and understand basics of traditional and non-traditional inheritance, understand the genetic and molecular mechanisms underpinning the phenotype variation, grasp the basis and limitations of conventional and next generation sequencing (NGS) genetic laboratory diagnosis, interpretation of genetic results with particular reference to pathogenicity

of NGS mutations or variants. New opportunities for genetic and genomic education are now made available through 'in house' formal and informal seminars, workshops and structured post graduate Diploma/ Masters level courses. The experience and outcomes of MDT based specialist healthcare have shown positive gains in the UK NHS. The UK 100,000 Genomes Project lays emphasis on genetic and genomic education directed at different level of the healthcare manpower for effective and efficient specialist healthcare delivery, particularly for rare inherited diseases.

### **Pharmacogenomics in Latin American Admixed Populations**

*Guilherme Suarez-Kurtz*

Brazilian Pharmacogenomics Network,  
Rio de Janeiro, Brazil

The heterogeneous Latin American population, with European, African and Amerindian ancestral roots provides a model case for exploring the impact of population admixture on the frequency distribution of polymorphisms in pharmacogenes, design and interpretation of pharmacogenomics trials. We used the Affymetrix Drug Metabolizing Enzymes and Transporters (DMET) Plus array to characterize the distribution of 1936 pharmacogenetic polymorphisms and the individual proportions of Native American,

African and European biogeographical ancestry in healthy adults from the two most populous Latin American countries, Brazil and Mexico. The Brazilian cohort was stratified into census categories of "Race/Color", as White, Brown and Black, whilst the sample from Mexico comprised Native American Zapotecas and self-identified Mestizo individuals from 5 geographically distant regions. A substantial variation in individual ancestral proportions across and within census categories in Brazil, and geographic regions in Mexico was observed. Analysis of the extent of genetic differentiation (measured as  $F_{ST}$  values) revealed a long tail of markers showing large frequency differences in genes belonging to the Cytochrome P450, Solute Carrier (SLC) and UDP-glucuronyltransferase (UGT) families as well as genes affecting drug pharmacodynamics, such as VKORC1. Non-linear piece-wise smooth logistic regression modeling revealed that the odds of having a variant of pharmacogenomic interest vary over ranges determined by the frequency of the variant in the relevant ancestral populations, i.e. mainly Europeans and Africans for Brazilians, and European and Native Americans for Mexicans. The larger the difference in frequency between the relevant parental populations, the more inappropriate it is to refer to a "Brazilian", "Mexican", or "Latino/Hispanic" allele frequency. Population admixture of Latin American adds complexity to, but also

creates advantages for pharmacogenomics research, including the opportunity to explore pharmacogenomic associations in individuals with heterogeneous genetic ancestry under similar environmental and socio-economical conditions, and to gather information on peoples that are excluded or under-represented in clinical drug trials. These opportunities will be illustrated with data for warfarin and tacrolimus in Brazilian patients.

### **More than regulatory challenges for genomic medicine in Latin America**

*JA Lecaros*

Director Observatory for Bioethics and Law

Regulatory gaps in genomic medicine in Latin America are only one of the reasons explaining the disparities and inequities in the provision of genetic services and in genetic research compared to the developed countries. The regulatory challenges of our region to improve genomic medicine must be taken with a collective view and not country by country. It is not enough to improve national legislation. It is also necessary to create an institutional framework to discuss, design and apply public health policies with effective participation of citizens, medical genetics professionals, researchers and other stakeholders relevant to the genomic medicine of

our region. Finally, the improvement of our legislation must take into account, at the time of its application, the relevant role played by intermediary bodies such as IRBs, genetic counselors and scientific and medical associations with their best practice guidelines. The idea is that the legislation be clear and precise but not too extensive, leaving in these intermediate bodies the case-by-case application of genetic rights.

### **Precision medicine initiative in China called international**

*Yixue Li*

CAS-MPG Partner Institute for Computational Biology (PICB); Shanghai Institutes for Biological Sciences (SIBS); Chinese Academy of Sciences (CAS)

Almost exactly one year after US President Barack Obama announced the Precision Medicine Initiative, China is finalizing plans for its own country. In this national Precision Medicine Initiative (PMI) project, huge amounts of clinical data, from genome sequences to health records will be produced to help determine how drugs affect people in different ways. One and half years past after announcement of PMI in China, a lot of Universities, hospitals, institutions and companies involved in the project and line up to produce, gather and analyze the data, some observers worry that

problems with the nation's health-care related infrastructures, especially the problems resulted from lacking of infrastructures of National Bio-Medical Database Systems. Most of people realize that China needs a national bio-medical database system to support PMI in China like those databases maintained by NCI, NCBI, EBI and ICGC which take the responsibilities to gather, manage, clean, curate, deliver and share the data. Here we give a short instruction to PMI in China, and some initiatives for setting up national infrastructure of Bio-Medical Database Systems. Finally, we would like to call international collaborations, collaborative projects and strategies that will help to design and set up Chinese National infrastructures of Bio-Medical Database System, serve the global community to share data, set up related standards and take genomic advances to improve clinical care.

### **Implementing Genomics in the Health Care System in Canada**

*Catalina Lopez Correa*

CSO Genome British Columbia

Canada in general, and the province of British Columbia in particular, have long recognized the potential of genomics and its applications in clinical practice and in the healthcare system overall. The Canadian Genomics Enterprise has made significant investments in genomics, at

a research and application levels, over the last 17 years. Canada now boasts a vibrant research and clinical innovation ecosystem including core genomic platforms, world leading research and clinical centers, patient registries and electronic medical records. All these investments are now paying off as genomics is finding its way to the clinic in BC and in Canada. Examples will be provided on the applications of genomics in the areas of cancer, rare diseases, infectious diseases and pharmacogenomics, the four areas recognized globally as the most “clinically ready” and most likely to benefit patients and to have a positive economic impact in the health care system. Aside from focusing on these therapeutic areas, Genome BC is also developing initiatives in four cross-cutting themes that have been identified as key elements to help advance clinical implementation of genomics: Education, Big Data, Capacity and Access.

### **The US Precision Medicine Initiative and Million Veteran Program**

*Teri Manolio*

Division of Genomic Medicine, National Human Genome Research Institute, Bethesda, MD

The Precision Medicine Initiative Cohort Program, recently renamed the “*All of Us*” Research Program, is an ambitious project to recruit and study

the health and medical care of one million or more volunteers reflecting the broad diversity of the U.S. *All of Us* will begin by recruiting participants from 8 regional medical centers, 6 Federally Qualified Health Centers, and roughly 20 sites in the Department of Veterans Affairs Million Veteran Program. Individuals can also sign up directly as “Direct Volunteers” and attend a local free-standing clinic for a brief examination. Data collection will begin with a limited set of standardized data from sources including questionnaires, electronic health records, a baseline physical evaluation, biospecimens, mobile/wearable technologies, and geospatial/ environmental data. Data types will grow and evolve as science, technology, and trust evolve. The program is expected to begin recruiting shortly and will take 3-4 years to recruit one million participants.

### **Accurate and Rapid WGS Interpretation with Fabric Genomics**

*Anthony Fejes\*, Anna Lewis\*, Charlene Son-Rigby\*, Edward Kiruluta\*, Mark Yandell\*\*, Martin G. Reese\**

\* Fabric Genomics; \*\* University of Utah

Laboratories, healthcare systems, and country projects all share a need for an intelligent, modular and powerful

compute infrastructure in order to manage large-scale genomic interpretation, analysis, and clinical reporting. These projects typically include research, data mining, and clinical healthcare reporting components that require a broad array of tools and supporting resources. Additionally, there is a need for the preservation of patient genetic data for future re-interpretation and clinical studies. At the core of a precision medicine project is genome interpretation, which has to demonstrate both clinical utility and accuracy to improve patient care. In this talk, we will discuss Fabric Genomics' Opal Clinical software platform, a scalable software-as-a-service (SaaS) solution for interpreting vast amounts of NGS data from panels, exomes, and genomes. We will highlight examples of genomic testing that are transforming medical care, such as the 100,000 Genomes Project in England and Rady Children's Institute for Genomic Medicine (Rady Children's). The 100,000 Genomes Project, spearheaded by Genomics England (GeL), is a country study aimed at identifying disease-causing genetic variants in patients and families with rare genetic diseases and cancer using WGS. Opal Clinical has provided GeL with potential causative candidates in 44.7% of cases. At the core of Opal Clinical are Fabric Genomics' leading algorithms VAAST and Phevor, which are gene ranking methods developed and published in

collaboration with the University of Utah. Another example that we will present will be from Dr. Stephen Kingsmore's clinical group in pediatrics at Rady Children's Hospital. That group has a goal of rapid genome sequencing and analysis, with a 24-hour turnaround time from blood sample to result for critically-ill pediatric patients. Fabric Genomics' Opal STAT version of Opal prioritizes extremely urgent pediatric cases, and guarantees the delivery of comprehensive annotations on whole genomic data in less than 1 hour. We will present scientific results from these examples, as well as infrastructure features that address many concerns of scaling large scale genome projects, including data storage, scalability, data access, integrations, and regulatory requirements.

### **The Clalit Israeli 100K Personalized Medicine RCT**

*Gad Rennert*

Director, Clalit National Personalized Medicine Program and National Cancer Control Center Professor and Chairman, Dept. Community Medicine & Epidemiology, Carmel Medical Center and B. Rappaport Faculty of Medicine, Technion

The genomic medicine approach is promoted as a game changer, potentially leading to better health

status of the population. The leading and overall aim of the planned trial is to test the value of incorporating personalized genetic information into routine clinical service. The study will be a randomized controlled trial where primary-care clinics will be randomized into intervention vs. usual care. Intervention will include education on genetics of the community and of the involved primary medical care teams, followed by enhanced exome testing of all insured of the intervention clinics and employment of clinical actions that stem from the genetic test results (mostly identification of disease risk-alleles and pharmacogenetic treatment/health behavior modifiers). Our hypothesis is that the intervention arm of the study will experience reduced morbidity/mortality rates, will show better disease control indicators, improved quality of life and improved economic balance. Major "byproducts" of the project will involve mapping the genetic background structure of Israeli Jewish and non-Jewish populations to serve as reference for future discovery and identification of genetic variants associated with specific diseases and with response to medications. 40 primary care clinics with 250,000 adults serviced by them will be randomized into 20 intervention clinics and 20 usual care clinics. 100,000 people are expected to consent to be tested in the intervention arm. Testing will involve

whole exome analysis supplemented with an extensive SNP-array (with DNA from buccal swabs) and potentially parallel testing of the microbiome using a stool sample.

### **Genomic Medicine in South America: current status**

*Gabriela Repetto, MD*

Center for Genetics and Genomics, Facultad de Medicina, Clinica Alemana Universidad del Desarrollo, Santiago, Chile

In order to assess the current status of implementation of Genomic Medicine in South America, a survey was sent to presidents of each country's Genetics Society and the Asociacion Latinoamericana de Genetica, requesting information on ongoing nationwide initiatives. Two countries have established databases on local genetic variation: [www.bipmed.com](http://www.bipmed.com) (Brasil) and [www.chilegenomico.cl](http://www.chilegenomico.cl). Three countries (Argentina, Ecuador, Uruguay) are developing platforms for high throughput sequencing. Disease-focused genomic initiatives are being carried out in Chile (lung cancer [www.cemp.cl](http://www.cemp.cl)) and Argentina (rare disorders [www.bitgenia.com](http://www.bitgenia.com)). There is an Iberoamerican Network on Pharmacogenetics and Pharmacogenomics ([www.ribef.com](http://www.ribef.com)), focused on collaborative research in the field. At the public policy level, Peru, Colombia and Argentina have laws that promote access to diagnosis



and care for individuals with rare disorders; the Ministry of Health in Chile has convened a group of experts to develop a road map for the inclusion of Genomic Medicine in the country. Although the survey did not identify nationwide programs already using Genomic Medicine in the clinical setting, all these initiatives are gradually building relevant and necessary capacities for its development. Collaboration between countries should be encouraged to move the field forward and to allow individual patients and countries to benefit from genomics-led health care.

### **Fostering Genomic Literacy for Today and Tomorrow**

*Laura Lyman Rodriguez*

Division of Policy, Communications & Education, NIH/NHGRI  
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If genomic medicine is to be optimally integrated into clinical care, and society more broadly, a certain level of genomic literacy is required across health provider teams, and importantly among patients and the general public (i.e., the future workforce and patients-to be). NHGRI has been focused on targeted outreach and educational support for students and health providers for some time, but with accumulating advances in genomic medicine approaches, the

urgency and scope of opportunities to be pursued has increased. This talk will review NHGRI initiatives to improve genomic literacy in the recent past and preview exciting new opportunities to build a collaborative network and national campaign in this area across the learning life-span (from K to gray).

### **From human genetic insights to drug discovery impact**

*Nadeem Sarwar*

President, Eisai Inc.

The opportunity provided by human genetics to disrupt the drug discovery process is now well recognized. Compelling data indicate that therapeutic targets supported by robust human genetic evidence are more likely to deliver effective new medicines than those that are not. Genetics and other sources of human biology data not only provide an opportunity for identification of novel drug targets more likely to work, but also for identification of patient sub-populations more likely to benefit. Despite this recognition, and despite considerable investment being made across sectors to harness the potential of human genetics and precision medicines, fully realizing a new paradigm in human genetics guided drug discovery requires three key changes to be implemented:

1. Removal of scientific silos to enable integration and innovation without borders
2. Supporting an ownership culture that rewards truth-seeking behavior
3. Entrepreneurial collaborative models that meet all stakeholders' needs

### **Australian Genomics Health Alliance: implementing genomics into health-care.**

*Andrew Sinclair*

Murdoch Children's Research Institute, Royal Children's Hospital and Dept. of Paediatrics, University of Melbourne, Melbourne, Australia

The Australian Genomics Health Alliance (AGHA) is a national network of clinicians, diagnosticians and researchers working towards the development of genomic medicine both within Australia, and in collaboration with international consortia. The AGHA is setting out a roadmap for the adoption of genomics into clinical care by undertaking research within four key programs of activity. Program 1: National diagnostic and translational research network; Program 2: Federated national genomic data repository; Program 3 Economics and policy development; Program 4: Genomic workforce education and ethics. The prospective collection and evaluation of data from patients recruited in each state within

two disease flagships in rare disease and cancer will drive strong national diagnostic networks and provide robust economic and clinical evidence on patient health outcomes and cost effectiveness. This will inform the development of educational, ethical and policy recommendations and sustainable funding pathways to implement genomic medicine into Australian healthcare.

### **Undiagnosed severe intellectual disability**

*Joris A. Veltman<sup>1,2</sup>*

<sup>1</sup>Institute of Genetic Medicine, Newcastle University, Newcastle, United Kingdom

<sup>2</sup> Department of Human Genetics, Radboud University Medical Center, Nijmegen, the Netherlands

Severe intellectual disability (ID, IQ<50) occurs in 0.5% of newborns and is largely genetic in origin. The extensive genetic heterogeneity of this disorder requires a genome-wide detection of all types of genetic variation. In the last 15 years we have developed and applied various genomics approaches, from genomic microarrays to exome and genome sequencing, to improve the diagnostic yield in this disorder. These studies revealed the major role of rare *de novo* point mutations, indels and CNVs in ID, requiring a patient-parent trio-based diagnostic approach. Recently, we

applied genome sequencing to 50 patients with severe ID and their unaffected parents. Notwithstanding extensive genetic prescreening, including microarray-based CNV studies and exome sequencing, a conclusive genetic diagnosis was reached in 20 patients. This results in a diagnostic yield of 42% in this extensively studied cohort, and 62% as a cumulative estimate in an unselected cohort.

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## **Genomic Test Evaluation Frameworks**

*Robyn Ward, AM FAHMS*  
Deputy Vice-Chancellor (Research)  
and Vice President (Research)

**Aims and Background:** The aim of this study was to examine the theoretical and practical frameworks used to assess genomic tests world-wide, drawing on a systematic literature

review and a survey of international stakeholders in genomic testing. While multiple different assessment frameworks exist, we find that no golden standard has been adopted and the majority of practical frameworks assessed share only some common assessment criteria while other are included on a tailor-made basis. These findings illustrate the challenges with which the assessment of genomic tests for reimbursement, regulatory and certification purposes grapples around the world, and indicates a need for a robust dialogue and sharing of lessons learned. Advances in genetic and genomic testing have led to a proliferation in the number and types of genetic tests available. However, large-scale genomic tests also have some potential downsides that are either absent or less of a concern for most other medical test types. For example, large-scale genomic tests can lead to unnecessary tests and treatment not only for the patient, but also for his or her genetic relatives. In addition, genome-scale tests can generate dozens of incidental findings.

**Observations:** Frameworks used to evaluate genetic and genomic tests often consider only benefits and risks common to most screening, predictive, diagnostic and prognostic medical tests and fail to explicitly consider risks that are specific large-scale genomic tests.

**Conclusions:** The ability of clinicians and reimbursement organisations to properly evaluate large-scale genomic

tests is lagging behind the implementation of the tests themselves. Such organisations could benefit from increased information sharing and standardisation of evaluation frameworks.

## **Public Health Pharmacogenomics**

***George P. Patrinos***

*Associate Professor of  
Pharmacogenomics and  
Pharmaceutical Biotechnology,  
University of Patras, Greece*

The central dogma of genomic medicine is to exploit an individual's genomic profile to support the clinical decision-making process and to individualize drug treatment modalities. Also, pharmacogenomics has a pivotal role in genomic medicine aiming to delineate drug efficacy and toxicity with the underlying genomic composition in genes involved in the pharmacokinetics and pharmacodynamics of drug regimens. There are various examples of the use of genome-guided therapeutic interventions in many medical specialties, mainly oncology and cardiology. Although it is obvious that there have been major leaps in pharmacogenomic research, facilitated by the genomic technology revolution, the pace of these discoveries have not met with reciprocal advances in the translation of these findings into the clinic, resulting into the

personalization of conventional therapeutic interventions. To this end, there are often significant barriers that hamper the smooth incorporation of pharmacogenomics research findings in the daily medical practice, which have to do more with disciplines related to Public Health Genomics rather than pharmacogenomics research itself. These disciplines touch upon pharmacogenomics in relation to Public Health Genomics disciplines, such as ethics in genomics, economic evaluation in genomic medicine, genome informatics and knowledgebases, and the involvement and genetics education of the various stakeholders in the field of pharmacogenomics, mostly healthcare professionals and the general public. In this lecture, results from various projects related to pharmacogenomics touching upon public health disciplines will be presented.

## **Undiagnosed Diseases Network International (UDNI)**

Domenica Taruscio <sup>1</sup>, S. C. Groft <sup>2</sup>, H. Cederroth <sup>3</sup>, B. Melegh <sup>4</sup>, P. Lasko <sup>5</sup>, K. Kosaki <sup>6</sup>, G. Baynam <sup>7</sup>, A. McCray <sup>8</sup>, G. Floridia <sup>1</sup>, M. Salvatore <sup>1</sup>, M.C. De Stefano <sup>1</sup>, W. A. Gahl <sup>2</sup>

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Undiagnosed diseases are a global health issue, calling for an international scientific and healthcare effort. In 2008, the National Institutes of Health (NIH) Undiagnosed Diseases Program (UDP) was initiated to provide diagnoses for individuals who had long sought one without success. Moreover, as a result of two international conferences (Rome 2014 and Budapest 2015), the Undiagnosed Diseases Network International (UDNI) was established, modeled in part after the NIH UDP and the recently formed US-wide Undiagnosed Diseases Network (UDN). The UDNI has published a consensus framework of principles, best practices and governance; the Board of Directors reflects its international character, as it includes experts from Australia, Canada, Hungary, Italy, Japan and the USA. The UDNI involves centers with internationally recognized expertise, and its scientific resources and know-how aim to fill the knowledge gaps that impede diagnosis. Consequently, the UDNI fosters the translation of research into medical practice. Active patient involvement is critical; the

Patient Advisory Group is expected to play an increasing role in UDNI activities. After the UDNI launch (2015), several countries have activated national Networks (e.g., Australia, Italy, Austria, Japan) operating in the framework of UDNI and in collaboration with other UDNI members, who convened in Vienna in February of 2016 and in Tokyo in November of 2016 to plan for data sharing. The coming Conference will be organised in Stockholm in August 30-31, 2017. All information for physicians and patients is available at the UDNI website (<http://www.udninternational.org>).

### **The Science May be the Easy Part: Challenges and Solutions in Implementing a Molecular Diagnostic Test**

*James Wingrove*  
CardioDx

Successful implementation of a commercial molecular diagnostic test requires the correct alignment of clinical, technical, commercial and financial initiatives. To be clinically useful, it is paramount the test informs and guides meaningful clinical decisions; in addition access to the correct biological samples coupled with precise clinical data and phenotyping is important when generating the clinical trial data necessary to develop and validate the

assay. From the technical side, platform choice including reliability, reproducibility and the ability to scale can have a large impact on throughput, turnaround time, and cost. Successful commercial adoption in the clinic requires an understanding of the customer, with a primary focus on the ordering clinician and how the test will integrate into their practice, as well as the impact of the test on cost savings for the managed care system. Finally,

the financial aspect of implementation can be challenging, with a potentially long pathway to viability requiring deep financial resources. Examples of these obstacles, and how they were overcome, will be illustrated using the development and commercial implementation of a peripheral blood gene expression test for the detection of obstructive coronary artery disease as an example.



# Global Genomic Medicine Collaborative

## ABSTRACTS – CONFERENCE PARTICIPANTS

### **The Application of Genomic Medicine in the UAE**

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The interplay between genomics and medicine led to the development of a new discipline in medicine called “Genomic Medicine”, which involves the use of genomic information about an individual as part of their clinical care. This discipline is already making a significant global impact in the fields of oncology, pharmacology, infectious diseases, rare and undiagnosed genetic conditions. The utility of genomic medicine is in providing: (1) highly accurate diagnostic tools (2) unprecedented predictive power of disease risk (3) opportunities for developing novel and personalized treatments and (4) new avenues for the development of creative

prevention strategies. In recent years, we have been using molecular, genomic and cellular approaches to elucidate the underlying genetic causes and mechanisms of single gene rare disorders in the United Arab Emirates (UAE). Collectively, those disorders are particularly highly prevalent among Arab populations, including the UAE, due to the high rates of consanguinity within these populations. Our research resulted in the identification of numerous mutations in many patients with recessive disorders and the elucidation of the cellular basis of several single gene disorders. In this article, I will present examples on our approach of using next-generation whole-exome sequencing for the identification of disease genes and mutations underlying recessive disorders among Emirati families. In addition, the elucidation of the cellular mechanisms underlying some single gene disorders will be presented. Furthermore, the areas of research and development needed to implement genomic

medicine in the UAE will be highlighted.

## **Systems Biology of Stem Cells**

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Embryonic stem (ES) cells are characterized by their ability to self-renew and remain pluripotent. Transcription factors have critical roles in the maintenance of ES cells through specifying an ES-cell-specific gene expression program. Deciphering the transcriptional regulatory network that describes the specific interactions of these transcription factors with the genomic template is crucial for understanding the design and key components of this network. To gain insights into the transcriptional regulatory networks in ES cells, we use chromatin immunoprecipitation coupled to ultra-high-throughput DNA sequencing (ChIP-seq) to map the locations of sequence specific transcription factors. These factors are known to play different roles in ES cell biology. Our study provides new insights into the integration of these regulators to the ES cell-specific transcription circuitries. Collectively, the mapping of transcription factor binding sites identifies new features of

the transcriptional regulatory networks that define ES cell identity. Using this knowledge, we investigate nodes in the network which when activated, will jump-start the ES cell-specific expression program in somatic cells.

## **Establishing a Global Genomics Nursing Alliance**

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Background: Nurses have a pivotal role in bringing the benefits of genomics and precision medicine to everyday healthcare. However, concerted global effort is needed to transform nursing policy and practice and address widely acknowledged deficits in nurses' genomic literacy. Aim: Increase nursing genomic literacy by gathering consensus from global leaders and nursing experts on key attributes of a Roadmap for action consisting of four complementary activities: 1. Building



international partnerships for collaboration, consultation, sharing, and research; 2. Establishing a Maturity Matrix to inform strategies to increase genomic literacy, improve care and benchmark progress that recognises real-world constraints; 3. Identifying existing nursing assets in genomics that could be shared throughout the international nursing community; 4. Determining key features needed for a Global Genomics Nursing Alliance (G2NA) platform to facilitate G2NA ongoing work. Methods: A 3-day invitational Retreat utilised Liberating Structures techniques in an innovative participatory approach to a structured programme of activities. Data were gathered prior to the invitational retreat through two online scoping surveys. Additional data was collected during the 3-day invitational Retreat utilizing real time voting through Turning Point. Outcomes: 29 participants from 17 countries and 7 major organisations attended. Common challenges around education and resources included access need for resources to link genetic variation to clinical implications (64%) and limited access to point of care educational information and clinical decision support (71%). All countries reported existence of genetic specialty services but only 4 reported nursing leadership driving genomic integration in nursing. Essential action areas based on prioritised needs were agreed as; 1. Enhanced education and workforce

development; 2. Effective nursing practice that builds on an evidence base, with clear delineation of nursing roles and interventions; 3. Infrastructure and resources to support education, practice and services; 4. Collaboration and communication across borders and professional groups; 5. Person, family and community-focused care; 6. Leadership in transforming healthcare through policy development. There was consensus (100%) that G2NA was an appropriate vehicle to increase genomic nursing capacity, guided by the Roadmap, and that it should seek to influence regulation and standards. Two workgroups have been established, strategic planning and funding to define G2NA operations. Additionally, expansion of the existing G2NA website ([www.g2na.org](http://www.g2na.org)) is underway, a G2NA listserv to facilitate communication has been implemented. Efforts to explore expansion of the Genetic/Genomic Competency Center for Education globally and G2NA inter-professional collaboration are underway. Conclusion: The initiative generated considerable social capital to accelerate change across global nursing communities.

#### **Minimum Information required for a DMET Experiment reporting**

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The DMET (Distribution, Metabolism, Excretion and Toxicity) technology has steadily advanced leading to increased demands to develop new bioinformatics software, analysis tools, algorithms, web applications and specific statistical techniques. With the advent of personalized medicine, it is evident that a large number of pilot studies will be conducted globally in the foreseeable future. Findings between different studies can be compared effectively if data reporting standards are in place. Furthermore, if data are extracted in a concise and correct manner, they can be used in subsequent experiments. We feel that quality of data supersedes quantity

and using a concise method of data extraction from DMET arrays can greatly increase the experimenter's ability to sort biologically meaningful information from background noise and experimental error. Ultimately, the field of pharmacogenomics will evolve to integrate data from various omics and technology platforms. Implementing standards for submission of data for each of the various platforms (microarrays, SNP arrays, proteomics and DMET), will aid the development of pipelines able to consolidate the different platforms and the standards by which each abides. Minimum Information required for a DMET Experiment (MIDE) is proposed to provide pharmacogenomics reporting guidelines, the information and tools required for reporting to public omic databases. For effective DMET data interpretation, sharing, interoperability, reproducibility and reporting, we propose MIDE.

### **Pharmacogenomics of adverse drug reactions in Thailand**

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In some regions, adverse drug reactions are disproportionately

affected the vulnerable populations. The fundamental to these unfortunate events are genetically mediated adverse drug reactions. The higher prevalence of the adverse drug reactions in Thailand came with the opportunity. The Recent implementation of rational drug use policy incorporated the basic concept of pharmacogenomics, with the recommendation to provide pharmacogenetic tests where the health service system has access to the pharmacogenomics services. With rapid cost reduction, the implementation of pharmacogenomics become an important policy that shall provide the immediate health benefit and cost savings to the health system.

### **The genetic basis of common conditions: the CHRIS study**

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The Cooperative Health Research In South Tyrol (CHRIS) study is a population-based study with a longitudinal lookout to investigate the genetic and molecular basis of age-related common chronic conditions and their interaction with life style and environment. Adults from an Alpine valley of South Tyrol are invited, with more than 9500 participants enrolled at the start of 2017 and 13,000 anticipated by the end of 2018. This population is characterized by long-term social stability and homogeneous environment which should both favour the identification of enriched genetic variants. Family participation is encouraged for complete pedigree reconstruction and disease inheritance mapping. Extensive pedigree reconstruction is possible given the structure of the population in the valley. Computer-assisted interviews have been implemented for health assessment of the cardiovascular, endocrine, metabolic, genitourinary, nervous and cognitive systems. Fat intake, cardiac health, and tremor are assessed instrumentally. Nutrient intake, physical activity, and life-course smoking are measured semi-quantitatively. Participants are phenotyped for 70+ blood and urine parameters, and cryo-preserved urine,

DNA, and whole and fractionated blood is biobanked. Metabolite profiling, both targeted and untargeted, is being assessed through liquid-chromatography mass-spectrometry analysis. Samples are genotyped at ~1 million variants using the Illumina HumanOmni ExpressExome array. The first data release including 4570 fully phenotyped and genotyped samples is now available for analysis. Whole exome sequencing of the first 4000 participants will be completed within 2017. A first follow-up is foreseen seven years after the initial baseline visit, with focused and general follow-up studies being planned in the future. The CHRIS study is producing a valuable resource to assess the contribution of genomics, metabolomics, and environmental factors to human health and disease. More specifically, it is a source of valuable data for the investigation of potential low-frequency/rare variants contributing to complex traits that are enriched in the target population. Moving forward we envisage the study will be contributing to the work of international consortia, and will provide a useful resource for integrating and testing tools aimed at integrating multi 'omics' data into programs aimed at improving preventative healthcare.

## **National Scale Precision Medicine in Singapore**

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In Nov 2016, Singapore established a National Precision Medicine Program (NPMP) with the mandate to coordinate an all-of-nation effort to transform the face of Singapore healthcare. Championed by the Ministry of Health and the National Medical Research Council, the NPMP bridges multiple funding agencies and research performer units, including A\*STAR, National Research Foundation, Economic Development Board, and Infocomm Media Development Authority among others. In this talk, we will describe our early progress in developing the NPMP, focusing on how this effort taps into ground-up efforts and lessons learned at the Singapore academic medical centers.

## **Implementing Pharmacogenomics in the Middle East developing**

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With the approval of pharmacogenetic testing by the FDA and the EMEA, medications have been recently used more safely and effectively, and the implementation of pharmacogenomics has become a fact. Further research is also ongoing to improve personalized patient medicine but mostly in developed countries. Pharmacogenomics information and implementation gap persists and has been highlighted in developing countries that are experiencing an exponential growth in chronic diseases; in 2020, it is estimated that 73% of deaths will be due to non-communicable diseases that can benefit from pharmacogenomics services if offered with local context to each developing country. The study aims at identifying the opportunities and challenges to overcome to contribute in making this approach feasible in the Middle East; Lebanon is a case study.

## **Using innovative education and training approaches**

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The success of the pioneering work of England's 100,000 Genomes Project is enabling the National Health Service (NHS) in England to routinely commission whole genome sequencing technologies from as early as 2018 in order to improve diagnosis, inform patient and family management and personalise treatment decisions in mainstream medicine, especially in the fields of rare disease and common cancer. The requirement for the rapid and widespread adoption of genomic medicine for the 55 million population of England requires innovative, flexible and accessible approaches to the education and training of the existing 1.3 million NHS workforce, as well as the prospective healthcare workforce. The National Genomics Education Programme was established with three aims: a) to upskill the existing highly specialised workforce to deliver the 100,000 Genomes Project through a programme of specific resource development across the project pipeline b) to build genomic capacity and capability in the current and prospective healthcare workforce

through the launch of new accredited professional training and academic curricula and courses including creating new specialisms such as clinical bioinformatics, and the development and funding of a multi professional Master's in Genomic Medicine and c) to support widespread adoption of genomics across the wider workforce through the development of communities of practice, including a network of training and education leads and the launch of a new NHS Faculty of Genomic Medicine. Using exemplars from across our extensive portfolio of work we will present data on resource uptake and learner experience to demonstrate the impact of our comprehensive and underpinning education and training framework for all healthcare professionals at all stages of the genomics education continuum for the rapid adoption of genomic medicine in the NHS. We will also discuss how this approach could be transferred to other international healthcare systems and populations.

### **Several HLA-B alleles are observed in carbamazepine induced**

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Several HLA-B alleles are observed in carbamazepine induced steven-johnson syndrome/ toxic epidermal necrolysis (SJS/TEN) patients in the Indonesian population. Carbamazepine (CBZ) is one of the most common causes of life-threatening cutaneous adverse drug reactions such as Stevens–Johnson syndrome (SJS) and toxic epidermal necrolysis (TEN). Previous studies have revealed a strong association between HLA-B\*15:02 and CBZ-induced SJS/TEN. This study is aimed to investigate which HLA-B alleles are commonly observed in the CBZ-induced SJS/TEN

patients in the Indonesian population. In total 9 cases of Carbamazepine-induced SJS/TEN were genotyped for HLA-B using Wakflow HLA typing kit combined with Luminex assay. About 77.78% HLA-B\*15:02 allele was found in the patient with CBZ-induced SJS/TEN. Additionally, we also found 18.18% of HLA-B\*15:21 allele, which is classified in the same group of B75 serotype with HLA B\*15:02 in the patient samples. In total, B75 were observed in 88.89% of samples. Interestingly, in this study we also observed the presence of HLA-B\*18:01

allele with frequency of 33.33%. The other allele in the same group with HLA- B\*18:01, HLA-B\*44:03 is also observed. Both alleles belong to B44 supertype (In total 55.56% of the samples). Our finding showed that not only HLA-B\*15:02, but also other HLA alleles in B75 serotype and B44 supertype can be the potential markers of Carbamazepine induced SJS/TEN in the Indonesian population. Further study is necessary to confirm the association between CBZ-induced SJS/TEN with the allele groups.







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