

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)

KEYNOTE SPEAKERS

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

SPEAKERS AND MODERATORS

Takeya Adachi (Tokyo, Japan)
Spiros Agathos (Urcuqui, Ecuador)
Fahd Al-Mulla (Safat, Kuwait)
François Bernier (Toronto, Canada)
Jeffrey Braithwaite (Melbourne, Australia)
Mark Caulfield (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 (Vancouver, 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)
Gabiella 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

3rd Global Genomic Medicine Collaborative Conference
Implementing Genomic Medicine into Practice

Organizer



Global Genomic
Medicine Collaborative

Local Organizer

The GoldenHelix
FOUNDATION

GoldenHelix[®]
SYMPOSIA

Sponsors



illumina[®]



Endorsed by



Genomic Medicine Alliance
Genomics & Personalized Medicine



Global Genomic
Medicine Collaborative

Implementing
Genomic Medicine
into Practice

Scientific Program

27-29
April
2017

DIVANI Apollon
Palace and Thalasso
Athens, Greece

DAY 1: APRIL 27, 2017

- 10:00 - 13:00 **Registration**
 13:00 - 13:10 **Welcoming 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*
George P. Patrinos, *Associate Professor of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras, Greece*
 13:10 - 13:25 **Welcome from Greek Genetics Society**
Aspasia Tsezou, *President, Hellenic Association of Medical Geneticists, Greece*
 13:25 - 13:55 **Keynote Lecture #1**
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

- 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)

- 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**

DAY 2: APRIL 28, 2017

- 08:15 **Opening 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*
 08:20 **Keynote Introduction**
George P. Patrinos, *Associate Professor of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras, Greece*
 08:25 **Keynote Lecture #2**
Stylianios Antonarakis, *Professor and Chairman of Genetic Medicine, University of Geneva Medical School, Switzerland*

Scientific Program

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

- 09:00 **Kuwait Genome Program**
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 **Implementing Genomic Medicine in 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

- 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, *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**

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

- 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, *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
 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)**

08:00 **Opening 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 of Research, and Vice President of Research, The University of Queensland, Australia*

SESSION VI: PHARMACOGENOMICS IN THE CLINIC: INTERNATIONAL PROGRESS

Moderator: George P. Patrinos, Associate Professor of Pharmacogenomics and Pharmaceutical Biotechnology, University of Patras, Greece

- 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, Associate Professor, Center of Pharmacogenomics and 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 **Pharmacogenomics 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, Ramathibidi Hospital, Thailand*
 09:40 **Panel discussion**
 10:15 - 10:30 **BREAK**
 10:30 - 12:00 **Breakout Reports and Discussion**
Moderator: Geoff Ginsburg, *Director, Duke Center for Applied Genomics & Precision Medicine, Professor of Medicine, Biomedical Engineering and Pathology, Duke University*
 12:00 - 13:00 **WORKING LUNCH [G2MC Steering Committee meeting (continued; 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

- 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**