

Functional Genomics Symposium

13-14 December 2015

Day 1 (13 December 2015)

07:00 - 08:00	Registration and refreshments
08:00 - 08:05	Opening remarks
	Professor the Lord Darzi of Denham PC KBE FRS
	Vice-Chairperson of the Board of Sidra Medical and Research Center
08:05- 08:10	Recent advances in genomic medicine in Qatar
	Francesco M Marincola, MD, FACS
	Chief Research Officer, Sidra Medical and Research
	Center, Qatar
Session 1	The international forefront of genomics and precision medicine
Moderator	Clare Turnbull
08:10 - 08:40	The Genomics England 100,000 Genomes project
	Clare Turnbull, PhD
	Clinical Lead for the Genomics England Cancer Program, Genomics England, UK
08:40 - 09:10	Genomes for medicine
	David Bentley, DPhil, FMedSci
	Vice president and Chief Scientist, Illumina Inc, USA
09:10 - 09:40	The Continuous spectrum of mutations spanning pediatric and adult disease
	Eric Boerwinkle, MD, PhD
	Kometsky Family Chair in Human Genetics, Professor and Director,
	Human Genetics Center and Department of Epidemiology, University of
	Texas, Health Science Center at Houston, School of Public Health, USA



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09:40 – 10:10	Population precision medicine: linking the clinic and research at scale Hákon Guðbjartsson, Phd Vice President of Bioinformatics and IT WuXi Next Code Genomics, USA
10:10 – 10:25	Enabling research by publishing human genomics Myles Axton, PhD Chief Editor, Nature Genetics, USA
10:25 – 10:45	Coffee break
Session 2	The need and readiness to implement genomic medicine locally
Moderator	Khalid Fakhro
10:45 – 11:15	Qatar Genome Program Asma Ali J F Al-Thani, PhD Head and Associate Professor of Virology, Health Sciences Department, College of Arts and Sciences, Qatar University Director of Biomedical Research Center, Qatar University Chair of Qatar Genome Program Committee - Board Vice Chairperson, Biobank, Qatar
	Wei Liu, PhD, Director of Clinical Genomic Lab, Sidra Medical and Research Center, Qatar Rashid Al Ali, PhD, Division Chief of Bioinformatics, Sidra Medical and Research Center, Qatar Ena Wang, MD, Division Chief of Translational Medicine, Sidra Medical and Research Center, Qatar
11:15 –11:45	The use of clinical exome sequencing in Middle Eastern patients with mendelian disorders Tawfeg Ben-Omran, MD, FRCPC, FCCMG Head, Clinical and Metabolic Genetics, Associate Professor, Weill Cornell Medical College, Department of Pediatrics, Hamad Medical Corporation, Qatar



11:45 - 12:15 Regulation in genetic testing and consultation **Eman Sadoun, MD** General Manager, Research Division, Healthcare Quality Management and Patient Safety Department, Supreme Council of Health, Qatar 12:15 - 12:45 Ethical issues in (Functional) genomics: Islamic perspectives Mohammed Ghaly, BA, MA, PhD Professor of Islam and Biomedical Ethics, Research Center for Islamic Legislation and Ethics (CILE), Faculty of Islamic Studies, Qatar 12:45 - 13:45 Lunch and poster viewing 13:15 - 13:35Lunch time session sponsored by Alliance Global Group Accurate Somatic mutation detection applied to liquid biopsy samples Jeff Jensen, Chief Executive Officer, Fluxion Biosciences, UAE Moderator **Tawfeg Ben-Omran** Functional genomics through the medical annotation of the human 13:45 - 14:15 genome Fowzan Alkuraya, MD, FAAP, FACMG Professor of Human Genetics, Principal Clinical Scientist/Senior Consultant, King Faisal Specialist Hospital and Research, Center, Saudi Arabia 14:15 - 14:45 Identification of De Novo and rare inherited DNA variants in autism spectrum disorder Hatem El-Shanti, MD Scientific Director, Qatar Biomedical Research Institute, Qatar 14:45 - 15:15 Exome sequencing in mendelian disease in Qatar Khalid Fakhro, PhD Principal Investigator, Congenital Disease Genetics Sidra Medical and **Research Center** Assistant Professor, Weill Cornell Medical College, Qatar



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15:15-15:45	Gene discovery in unique populations Marios Kambouris, PhD, FACMGG Senior Clinical Genetics, Acting Head, Pathology Genetics, Sidra Medical and Research Center, Qatar
15:45 - 16:00	Coffee Break
Session 3	Challenges and accessibility from big data to personalized medicine
Moderator	Ena Wang
16:00 - 16:30	Pancancer genome analysis and the path towards women's personalized medicine Xavier Estivill, MD, PhD Chair-Genetics Program, Experimental Genetics Division, Sidra Medical and Research Center, Qatar
16:30 - 17:00	Variation annotation and interpretation: tools and public data Gabe Rudy VP Product & Engineering, Golden Helix, Inc, Bozeman, MT, USA
17:00 - 17:30	Data-driven enablement of the hippocratic oath in a clinical oncology setting David B Jackson, PhD Chief Innovation Officer, Molecular Health GmbH, Germany



Day 2 (14 December 2015)

Session 4	Genomics guided healthcare
Moderator	Paolo Gasparini
07:00 - 08:00	Registration and refreshments
08:00-08:30	Expanding the number of hospitals practicing genomic medicine Howard Jacob, PhD
	Faculty investigator, Executive Vice President Genomic Medicine, Chief Genomic Medicine Officer,
	Hudson Alpha Institute for Biotechnology, Alabama, USA
08:30-09:00	From genetic discovery to gene yherapy- 30 Years of Experience Paolo Gasparini, PhD
	Division Chief of Experimental Human Genetics, Sidra Medical and Research Center, Qatar
09:00 - 09:30	The use of genetics isolates to dissect complex quantitative traits Nicola Pirastu, PhD
	Investigator, Sidra Medical and Research Center, Qatar
09:30 - 10:00	Highly multiplexed single molecule counting: NanoString platform from lab to clinic
	Alessandra Cesano, MD, PhD Chief Medical Officer, NanoString Technologies, USA
10:00 - 10:30	Coffee Break
Moderator	Marios Kambouris
10:30 - 11:00	Genomics in clinical practice: Lessons from the bedside Livija Medne, MS, LCGC
	Co-Director, Individualized Medical Genetics Center Division of Human Genetics, The Children's Hospital of Philadelphia, USA
11:00 - 11:30	Intrauterine molecular therapy for the fetus Jerry Chan Kok Yen, MD
	Research Director, KK Women's and Children's Hospital, Singapore



11:30 - 12:00 3D Genome organisation and transcription regulation Yijun Ruan, PhD Professor and Director of Genomic Sciences, The Jackson Laboratory for Genomic Sciences, USA 12:00 - 13:15 Lunch and poster viewing 12.15-12.35 Lunch time session sponsored by LabWare Arabia Streamlining data management and workflows for biomedical and clinical research labs Abdallah Maazi, General Manager LabWare Arabia, UAE **Session 5** Functional genomics and award abstract presentation Moderator **Yijun Ruan** 13:15 - 13:45 Gene discovery leads to new targeted therapies in congenital immunological diseases Michael Lenardo, MD Chief, Molecular Development Section and Director, NIAID Clinical Genomics program, National Institutes of Health, USA Transcriptome wide analysis of non-coding RNA function in zebrafish 13:45 - 14:15 Sridhar Sivassubu, PhD Senior Scientist, CSIR Institute of Genomics and Integrative Biology, India 14:15 - 14:45 Short stories on long non-coding RNAs Vinod Scaria, MBBS, PhD Scientist, CSIR Institute of Genomics and Integrative Biology (CSIR-IGIB), India 14:45 - 15:15 Next Generation epigenetics - Innovative research tools for accurate analysis of DNA modifications Hanna Schutz, PhD

Business Development Associate, Cambridge Epigenetix Ltd, UK



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Abstract awards and closing

15:15 – 15:45	Coffee Break
15:45 - 16:00	Novel genes for intellectual disability identified using whole genome sequencing and pathway analysis Farah R Zahir, Canada's Michael Smith Genome Sciences Center, Vancouver, BC, Canada
16:00 - 16:15	High altitude adaptive EPAS1 haplotype in the Himalayas Qasim Ayub, The Wellcome Trust Sanger Institute, Wellcome Genome Campus, UK
16:15 – 16:30	Abstract Award oral presentation (to be confirmed)
16:30 - 16:45	Abstract Award Ceremony
16:45 – 17:00	Closing Remarks Ena Wang, MD Division Chief of Translational Medicine Sidra Medical and Research Center, Qatar