

Functional Genomics 2018

8-10 December | QNCC | Doha, Qatar

Big Data to Clinic

Program Schedule

Conference Objectives:

- Understand the latest scientific advances regarding the collection and analysis of big data in the field of genomics and precision medicine.
- Gain oversight regarding the assessment of the functional consequences of variants in the human genome that appear to be associated with clinical phenotype.
- Have an appreciation of the translation of genomic knowledge in the prevention, diagnosis and treatment of rare and common genetic disorders of infancy.

Day 1: Saturday 8 December 2018 "Human Genomes and Big Data"

07:00 – 7:40 Registration and refreshments

Introduction:

Objective: Define the Aims of the Symposium and its Framework with Respect to Qatar and Sidra Medicine.

07:40 – 07:45 **Welcome note**
Mr. Peter Morris
CEO, Sidra Medicine - Qatar

07:45 – 07:55 **Overview of Sidra Research Achievements**
Dr. Christof Von Kalle
CRO, Sidra Medicine - Qatar

07:55 – 08:15 **The Functional Genomics Symposium Series**
Professor Xavier Estivill
Co- Chair of the Functional Genomics Symposium 2018

Session 1: The Message of Genes

Moderators: Nicole Soranzo and Jithesh Puthen

Objective: To Show the Bulk of Information that the Genome Contains and the Challenges to Decode the Message for Clinical Purposes.

8:15 – 8:45 **"From Genomics To Therapeutics: Uncovering And Manipulating The Genetic Circuitry of Human"**
Manolis Kellis
MIT Computer Science, CSAIL, Broad Institute- Massachusetts, USA

8:45 – 9:15 **"Tohoku Medical Megabank Project- A National Challenge to Realize Personalized Medicine"**
Masayuki Yamamoto
Tohoku Medical Megabank Organization- Sendai, Japan

9:15 – 9:45 **"Using Single-cell Genomics to Understand Cell fate Decisions in Early Mammalia Development"**
John Marioni
University of Cambridge- Cambridge, UK

9:45 – 10:00 **"Talk Title "**
Bernice Lo
Sidra Medicine- Doha, Qatar

10:00 – 10:15 **"Short Talk "**
Abstract Winner
Institution- Country

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| 10:15 – 10:30 | “Short Talk ” Abstract Winner Institution- Country |
| 10:30 – 11:00 | Coffee break |
| Session 2: | Genome Sequencing in Action |
| Moderator: | Manolis Kellis and Stephan Lorenz |
| Objective: | To Show how the Activities in Genome Sequencing Projects Define the Dissection of Human Phenotypes |
| 11:00 – 11:30 | “Genome Sequencing and Multi-Omics Phenotyping in Precision Medicine” Nicole Soranzo Wellcome Trust Sanger Institute- Hinxton, UK |
| 11:30 – 12:00 | “Long-Read Sequencing of Complex Genomes and Improved Structural Variation Characterization” Evan E. Eichler University of Washington School of Medicine- Seattle, USA |
| 12:00 – 12:30 | “The Use of Population Biobanks for Translational Research” Cisca Wijmenga University Medical Center Groningen- Groningen, NL |
| 12:30 – 12:45 | “Talk Title ” Donald Love Sidra Medicine- Doha, Qatar |
| 12:45 – 13:00 | “Short Talk ” Abstract Winner Institution- Country |
| 13:00 – 13:15 | “Short Talk ” Abstract Winner Institution- Country |
| 13:15 – 14:45 | Lunch break |
| Session 3: | Qatar Genome and Medical Knowledge |
| Moderators: | Asmaa Al-Thani and Evan Eichler |
| Objective: | To Present the Objectives and First Results of the Qatar Genome Program Analysis |
| 14:45 – 15:05 | “Title of Talk” Said Ismail Qatar Genome Proram- Doha, Qatar |
| 15:05 – 15:25 | “Title of Talk” Xavier Estivill Sidra Medicine- Doha, Qatar |
| 15:25 – 15:45 | “The Genetic Architecture of Health and Disease-related Trait in Qatari Population” Omar Albagha Hamad Bin Khalifa University- Doha, Qatar |
| 15:45 – 16:05 | “Title of Talk” Jithesh Puthen Sidra Medicine- Doha, Qatar |
| 16:05 – 16:20 | “Short Talk ” Abstract Winner Institution- Country |
| 16:20 – 16:35 | “Short Talk ” Abstract Winner Institution- Country |
| 16:35 – 16:50 | “Short Talk ” Abstract Winner Institution- Country |
| 16:50 – 17:00 | Conclusion of First Day |

Day 2: Sunday 9 December 2018
“Modelling and Treating Disease”

07:15 – 8:15 Registration and Refreshments

Session 4: Modelling Human Disease

Moderators: Xavier Estivill and Cisca Wijmenga

Objective: To Show how to Develop and Analyze Human Mutations in Animal Models

8:15 – 8:45 **“Title of Talk”**
Nicholas Katsanis
 Duke University- NC, USA

8:45 – 9:15 **“Liver Organoids for the Study of Human Biology and Disease”**
Meri Huch
 University of Cambridge- Cambridge, UK

9:15 – 9:45 **“ZECARDIO: A Versatile Screening Platform for Cardiovascular Functional Genomics and Drug”**
Javier Terriente
 ZeClinics, Drug Discovery Through Zebrafish- Barcelona, Spain

9:45:00 – 10:00 **“Short Talk ”**
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10:00 – 10:15 **“Short Talk ”**
 Abstract Winner
 Institution- Country

10:15 – 10:030 **“Short Talk ”**
 Abstract Winner
 Institution- Country

10:30 – 11:00 **Coffee Break**

Session 5: Exploring the Consequences of Mutation in Model Systems

Moderators: Tomo Kino and Anne Ferguson-Smith

Objective: To Show how to Evaluate the Consequences of Mutations in Model Systems

11:00 – 11:30 **“RNA Toxicity in Huntington’s Disease: A Therapeutic Target in Polyglutamine Disorders”**
Eulalia Marti Puig
 Institutiion- Country

11:30 – 12:00 **“Non-self Mutations: Neurodegenerative Diseases have Genetic Hallmarks of Autoinflammatory Disease”**
Robert Richards
 University of Adelaide- Adelaide, AU

12:00 – 12:30 **“Gene Discovery Mediated by the Drosophila Model Organism Screening Center”**
Hugo Bellen
 Baylor College of Medicine- Texas, USA

12:30 – 12:45 **“Short Talk ”**
 Abstract Winner
 Institution- Country

12:45 – 13:00 **“Short Talk ”**
 Abstract Winner
 Institution- Country

13:00- 13:15 **“Short Talk ”**
 Abstract Winner
 Institution- Country

13:15- 14:45 **Lunch Break**

Session 6: Sidra Medicine Lecture in Human Genetics

Moderators: Donald Love

Objective: To Show how to Develop and Analyze Human Mutations in Animal Models

15:00 – 15:40 **“Genetic Approaches to Therapy for Duchenne Muscular Dystrophy”**
Kay Davies
 Oxford Neuromuscular Centre- Oxford, UK

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| 15:40 – 16:50 | Women in Science Workshop |
| Objective: | To Show how Successful Careers in Science can be Achieved by Women |
| Panelists: | <ul style="list-style-type: none"> ▪ Kay Davies Professor of Genetics Oxford Neuromuscular Centre - Oxford, UK ▪ Elizabeth Phimister Deputy Editor The New England Journal of Medicine- Boston, USA ▪ Bernice Lo Investigator, Sidra Medicine- Doha Qatar ▪ Mariam Al Maadeed Vice President for Research & Graduate Studies, Qatar University- Doha, Qatar ▪ Kholoud Al- Shafai HBKU PhD Student, Sidra Medicine- Doha Qatar |
| 16:50-17:00 | Conclusion of Second Day |
| Day 3: Monday 10 December 2018 “Mother and Child Health” | |
| 07:15 – 8:15 | Registration and Refreshments |
| Session 7: | Mother and Child Health |
| Moderators: | Nicholas Katsanis and Luis Saraiva |
| Objective: | To Evaluate the Impact of Knowledge in Cellular Models in Managing Human Disease |
| 08:15 – 08:45 | “Investigating Cellular Fate decision in Mouse Embryos by Single-cell RNA- Sequencing” Antonio Scialdone Helmholtz Zentrum München- Munchen, Germany |
| 08:45 – 09:15 | “Genetic Regulators of Maternal Immune Tolerance and Pregnancy Success” Sarah Robertson Robinson Research Institute- Adelaide, AU |
| 09:15 – 9:45 | “New Applications of Genomic Medicine in Women's Health” Lee Schulman Feinberg School of Medicine of Northwestern University- Illinois, USA |
| 09:45 – 10:00 | “Short Talk ” Abstract Winner Institution- Country |
| 10:00 – 10:15 | “Short Talk ” Abstract Winner Institution- Country |
| 10:15 – 10:30 | “Short Talk ” Abstract Winner Institution- Country |
| 10:30 –11:00 | Coffee Break |
| Session 8: | Diagnosis of the Future Child |
| Moderators: | Khalid Fakhro and Meritxell Guarch |
| Objective: | To Show how to Evaluate the Consequences of Mutations in Genes that Lead to Human Genetic Diseases |
| 11:00 – 11:30 | “Variable Silencing of the Repeat Genome - Implications for Non-Genetic Inheritance” Anne Ferguson-Smith University of Cambridge- Cambridge, UK |
| 11:30 – 12:00 | “The Renaissance of Genomic Medicine” Stylianos Antonarakis University of Geneva Medical School- Geneva, Switzerland |
| 12:00 – 12:30 | “The Results of the Undiagnosed Rare Disease Program of Catalonia (URDCat)” Luis Perez-Jurado Pompeu Fabra University Barcelona- Spain and University of Adelaide- Australia |
| 12:30 – 12:45 | “Short Talk ” Abstract Winner Institution- Country |

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| 12:45 – 13: 00 | “Short Talk ” Abstract Winner Institution- Country |
| 13:00 – 13: 15 | “Short Talk ” Abstract Winner Institution- Country |
| 13:15- 14:45 | Lunch Break |
| Session 9: | Patient Genome Sequencing |
| Moderators: | Stylios Antonarakis and Bernice Lo |
| Objective: | To Show how Personal Genome Sequencing Provides Key Information to Manage Disease |
| 14:45 – 15:15 | “Clinical Genomic: Discovery and Building Genetics Models for Complex Traits” James R. Lupski Baylor College of Medicine- Texas, USA |
| 15:15 – 15:45 | “Title of Talk” Michael Lenardo National Institute of Allergy and Infectious Diseases (NIH)- Maryland, USA |
| 15:45 – 16:00 | “A Nuts and Bolts Approaches to Diagnosis of SCID Patients in the Era of Newborn Screening” Medhi Adeli Hamad Hospital- Doha, Qatar |
| 16:00 – 16:15 | “Short Talk ” Abstract Winner Institution- Country |
| 16:15 –16:30 | “Short Talk ” Abstract Winner Institution- Country |
| 16:30 –16:55 | Conclusion of Third Day |
| 16:40 –16:55 | Poster Awards |
| 16:55 – 17:00 | Closing Remarks |