At the end of the Conference, the participants will be able to:

- Identify diseases or disorders discussed which may benefit from genetic diagnosis or Precision Medicine.
- List examples of targeted or advanced therapeutic options for genetic diseases discussed.
- Name diseases and disorders that may be present in the Qatari population.

Functional Genomics 2018 Target Audience

- Physicians
- Nurses
- Pharmacists
- Allied Health

(Involved in the field of Biology, Genomics, Genomic Medicine, Genetics, Cancer and other Chronic Diseases.)

SPC STATEMENT

The Scientific Planning Committee has reviewed all disclosed financial relationships of speakers, moderators, facilitators and/or authors in advance of this CPD activity and has implemented procedures to manage any potential or real conflicts of interest.

QCHP STATEMENT

This Activity is an Accredited Group Learning Activity (Category 1) as defined by the Qatar Council for Healthcare Practitioners-Accreditation Department and approved for a maximum of 22 hours.

Program Schedule

Day 1: Saturday 8 December 2018 "Human Genomes and Big Data"	
07:00 – 7:40	Registration and refreshments
Introduction: **Objective:** **1. Able to discuss the role of Sidra Research and Functional Genomics for healthcare in Qatar.** **2. Describe the achievements of Sidra Research.**	
07:40 – 07:45	**Welcome note** Peter Morris CEO, Sidra Medicine - Qatar
07:45 – 07:55	**Overview of Sidra Research Achievements** Christof Von Kalle CRO, Sidra Medicine - Qatar
07:55 – 08:15	**The Functional Genomics Symposium Series** Xavier Estivill Co- Chair of the Functional Genomics Symposium 2018
Session 1: The Message of Genes **Moderators: Nicole Soranzo and Puthen Jithesh** **Objective:** **1. Describe lessons learned from large genome projects** **2. List examples of challenges encountered in decoding the genome for clinical purposes** **3. Explain advantages of population biobanks and big data projects**	
8:15 – 8:50	**"From Genomics To Therapeutics: Uncovering And Manipulating The Genetic Circuitry of Human"** **Manolis Kellis** MIT Computer Science, CSAIL, Broad Institute, Massachussetts, USA
8:50 – 9:25	**"Tohoku Medical Megabank Project- A National Challenge to Realize Personalized Medicine"** **Masayuki Yamamoto** Tohoku Medical Megabank Organization, Sendai, Japan
9:25 – 10:00	**"The Use of Population Biobanks for Translational Research"** **Cisca Wijmenga** Groningen University Medical Center, Groningen, Netherlands
10:00 – 10:15	**"Role of allele-specific RNA expression in human traits"** **Gaurav Thareja** Weill Cornell Medicine, Qatar
10:15 – 10:30	**"Incidence, Clinical Spectrum and Molecular Mechanisms of Permanent Neonatal Diabetes Mellitus in the State of Qatar"** **Sara Al-Khawaga** Sidra Medicine, Qatar
10:30 – 11:00	Coffee break
Session 2: Genome Sequencing in Action **Moderator: Manolis Kellis and Stephan Lorenz** **Objective:** **1. Discuss how the activities in Genome Sequencing Define the Dissection of Human Phenotypes.** **2. Explain the advantages of long-read sequencing of genomes** **3. Describe how multi-Omics approaches can be utilized for Precision Medicine**	
11:00 – 11:35	**"Genome Sequencing and Multi-Omics Phenotyping in Precision Medicine"** **Nicole Soranzo** Wellcome Trust Sanger Institute, Hinxton, UK
11:35 – 12:10	**"Long-Read Sequencing of Complex Genomes and Improved Structural Variation Characterization" *** **Evan E. Eichler** University of Washington School of Medicine, Seattle, USA
12:10 – 12:45	**"Investigating Cellular Fate Decision in Mouse Embryos by Single-cell RNA-Sequencing"** **Antonio Scialdone** Helmholtz Zentrum München, Munich, Germany
12:45 – 13:00	**"Exomes in a Clinical Setting: The Promise and the Delivery"** **Donald Love** Sidra Medicine, Doha, Qatar

13:00 – 13:15	**"Sidra's Pediatric Precision Medicine Program – Opportunities for Discovery and Collaborations"** **Khalid Fakhro** Sidra Medicine, Doha, Qatar
13:15 – 14:45	**Lunch break**
Session 3: Qatar Genome and Medical Knowledge **Moderators: Asmaa Al-Thani and Richard O'Kennedy** **Objective:** **1. Able to explain the objectives of the Qatar Genome Program (QGP)** **2. Name of diseases and disorders discussed that may be present in the Qatari population** **3. Describe lessons gleaned from the first results of the QGP Analysis**	
14:45 – 15:05	**"Overview- Qatar Genome Project"** **Said Ismail** Qatar Genome Proram, Doha, Qatar
15:05 – 15:25	**"The Biomedical Landscape of Genetic Variation in the Population of Qatar"** **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	**"Exploring Pharmacogenetic Variants in the Qatari Population"** **Puthen Jithesh** Sidra Medicine, Doha, Qatar
16:05 – 16:20	**"Extended Blood Group and Platelet Phenotype Prediction from Whole Genome Sequencing and Its Impact on RBC Transfusion-Related Alloimmunization in the State of Qatar"** **Zohreh Tatari- Calderone** Sidra Medicine, Doha, Qatar
16:20 – 16:35	**"Rare Diseases in Qatar: Genomics and Proteomics of Heritable Muscle Disorders"** **Alice Abdelaleem** Weill Cornell Medicine, Doha, Qatar
16:35 – 16:50	**"Genetics and Epigenetics linked to T2D functional pathways in Qataris"** **Noha A. Yousri** Weill Cornell Medicine, Doha, Qatar
16:50 – 17:05	**"In Vivo and in Silico Models for Qatari Specific Classical Homocystinuria as basis for Development of Novel Therapies"** **Gheyath Khaled Nasrallaha** Qatar University, Doha, Qatar
17:05 – 17:20	**"Interactome mapping using All-vs-All sequencing (AVA-seq) method"** **Nayra M. Al-Thani** Weill Cornell Medicine, Doha, Qatar
17:20 – 17:30	**Summary of First Day by Jithesh Puthen**
Day 2: Sunday 9 December 2018 **"Modelling and Treating Disease"**	
07:15 – 8:15	Registration and Refreshments
Session 4: Modelling Human Disease **Moderators: Luis Saraiva and Cisca Wijmenga** **Objective:** **1. Explain how functional genomics has been utilized for drug discovery** **2. Name the advantages of using organoids in the study of human biology and disease** **3. Describe how animal models can be used to study human disease variants**	
8:15 – 8:50	**"The Use of Functional Genomics to Accelerate Drug Discovery"** **Nicholas Katsanis** Duke University, North Carolina, USA
8:50 – 9:25	**"Long noncoding RNAs: A Potential Gold Mine of New Disease Genes"** **Rory Johnson** University of Bern, Bern, Switzerland
9:25 – 10:00	**"Liver Organoids for the Study of Human Biology and Disease"** **Meritxell Huch** University of Cambridge, Cambridge, UK
10:00 – 10:35	**"Organoid co-culture system to study gut brain signaling"**

	Diego V. Bohórquez Duke University, North Carolina, USA
10:35 – 10:50	**"Hypertrophic cardiomyopathy-linked variants of cardiac myosin binding protein C3 display altered molecular properties and actin interaction"** **Sahar Da'as** Sidra Medicine, Doha, Qatar
10:50 – 11:20	**Coffee Break**
Session 5: Exploring the Consequences of Mutation in Model Systems **Moderators: Edward Stuenkel and Anne Ferguson-Smith** **Objective:** **1. List the advantages of utilizing animal models for gene discovery** **2. Describe more about the impact of non-self mutations in human disease** **3. Explain what spatial transcriptomics can reveal about biology**	
11:20 – 11:55	**"RNA Toxicity in Huntington's Disease: A Therapeutic Target in Polyglutamine Diseases"** **Eulalia Marti-Puig** University of Barcelona, Barcelona, Spain
11:55 – 12:30	**"Non-self Mutations: Neurodegenerative Diseases have Genetic Hallmarks of Autoinflammatory Disease"** **Robert Richards** University of Adelaide, Adelaide, Australia
12:30 – 13:05	**"Gene Discovery Mediated by the Drosophila Model Organism Screening Center"** **Hugo Bellen** Baylor College of Medicine, Texas, USA
13:05 – 14:30	**Lunch Break**
14:30 – 15:05	**"When exomes are not enough: Approaches to working through unresolved neurological cases"** **Elizabeth Ross** Weill Cornell Medicine, New York, USA
15:05 – 15:20	**"Spatial transcriptomics of olfactory receptors for high throughput mapping of olfactory bulb glomeruli"** **Kevin Zhu (Travel Award)** Duke University, North Carolina, USA
Session 6: Inaugural Sidra Medicine Plenary Lecture **Moderators: Donald Love** **Objective:** **1. Describe what is Duchenne Muscular Dystrophy (DMD) and some of the challenges of DMD treatment** **2. List a few of the current DMD therapeutic approaches** **3. Discuss insights and advice on how to excel as a female in science**	
15:20 – 16:05	**"Genetic Approaches to Therapy for Duchenne Muscular Dystrophy"*** **Dame Kay Davies** Oxford Neuromuscular Centre, Oxford, UK
16:05 – 17:25 **Women in Science Workshop** Moderator : Bernice Lo and Kholoud Al- Shafai **Objective: Able to demonstrate to all Women scientists of Qatar how they can build up a successful scientific career from women who achieved top level positions in the scientific world.** **Panelists:** ▪ **Dame Kay Davies** Professor of Genetics Oxford Neuromuscular Centre, Oxford, UK ▪ **Elizabeth Ross** Professor of Neurology and Neuroscience Weill Cornell Medicine, New York, USA ▪ **Elizabeth Phimister** Deputy Editor The New England Journal of Medicine, Boston, USA ▪ **Souhaila Al Khodor** Investigator Sidra Medicine, Doha, Qatar ▪ **Mariam Ali Al Ali Al Maadeed** Vice President for Research & Graduate Studies	

	Qatar University, Doha, Qatar
17:25-17:30	**Conclusion of Second Day by Xavier Estivill**
Day 3: Monday 10 December 2018 "Mother and Child Health"	
07:15 – 8:15	Registration and Refreshments
Session 7: From Models of Cells to Disease Phenotypes **Moderators: Nicholas Katsanis and Khalid Fakhro** **Objective:** **1. Discuss lessons in the development of embryos learned from RNA sequencing** **2. Describe genetic regulators involved in maternity/pregnancy** **3. List technologies or approaches used for prenatal/carrier screening**	
08:15 – 08:50	**"Genetic Regulators of Maternal Immune Tolerance and Pregnancy Success"** **Sarah Robertson** Robinson Research Institute, Adelaide, Australia
08:50 – 09:25	**"New Applications of Genomic Medicine in Women's Health"** **Lee Schulman** Feinberg School of Medicine of Northwestern University, Illinois, USA
09:25 – 09:55	**"Pathologic Glucocorticoid Receptor Mutations: From their Clinical Manifestations to Structural Impact on the Glucocorticoid Receptor Protein Toward Future Tailored Treatment Using Modified Glucocorticoid Ligands"** **Tomoshige Kino** Sidra Medicine, Doha, Qatar
09:55 – 10:20	**"The LINC Complex is Essential for Gametogenesis"** **Henning Horn** Hamad Bin Khalifa University, Doha, Qatar
10:20 – 10:35	**"Immune Functional Studies of a Novel Pathogenic STK4 Genotype: A Case Report"** **Andrea Guennoun** Sidra Medicine, Doha, Qatar
10:35 – 10:50	**"Characterization of the Microbial Diversity in the Milk of Mexican Women"** **Karina Corona Cervantes (Travel Award)** Genetics and Molecular Biology Department, Mexico City, Mexico
10:50 – 11:20	**Coffee Break**
Session 8: Diagnosis of the Future Child **Moderators: Evan Eichler and Meritxell Huch** **Objective:** **1. Describe implications of epigenetics on human biology** **2. Discuss lessons gleaned from the Undiagnosed Rare Disease Program of Catalonia** **3. Discuss the findings from the noninvasive prenatal testing study at HMC**	
11:20 – 11:55	**"Variable Silencing of the Repeat Genome - Implications for Non-Genetic Inheritance"** **Anne Ferguson-Smith** University of Cambridge, Cambridge, UK
11:55 – 12:30	**"The Renaissance of Genomic Medicine"** **Stylianos Antonarakis** University of Geneva Medical School, Geneva, Switzerland
12:30 – 13:05	**"The Results of the Undiagnosed Rare Disease Program of Catalonia (URDCat)"** **Luis Perez-Jurado** Pompeu Fabra University, Barcelona, Spain, and University of Adelaide, Australia
13:05 – 13: 20	**"A Validation Study: NIPT to Identify Pregnancies at high Risk for Aneuploidies Using NGS Platform with Minimum of 2% Fetal Fraction"** **Sarmad Ali Ghulam Shabir** Hamd Medical Corporation, Doha, Qatar
13:20 – 14:45	**Lunch Break**

Session 9: Patient Genome Sequencing

Moderators: Stylianos Antonarakis and Bernice Lo

Objective:

1. **Describe how molecular diagnosis and basic research findings can lead to novel therapies for Precision Medicine**
2. **Distinguish between the molecular methods/assays for different types of mutations**
3. **Describe the importance of and role of a genetic counselor in genomic research studies**

14:45 – 15:20	**"Genomics Research Uncovers the Primary Role of Complement Pathway in a Familial Form of Protein Losing Enteropathy"** **Ahmet Ozen** Marmara University, Istanbul, Turkey
15:20 – 15:55	**"Returning Results from Genomic Research: NIAID'S Experience"** **Leila Jamal** National Institute of Health, Bethesda, Maryland, USA
15:55 – 16:30	**"A Nuts and Bolts Approaches to Diagnosis of SCID Patients in the Era of Newborn Screening"** **Mehdi Adeli** Sidra Medicine and Hamad Medical Corporation, Doha, Qatar
16:30 – 16:45	**Mutation in the GTPase GIMAP6 Leading to Reduced Autophagy in a Severely Immune Deficient Patient"** **Brittany Chao (Travel Award)** Oxford University, Oxford, UK
16:45 – 17:20	**"Clinical Genomic: Discovery and Building Genetics Models for Complex Traits"** **James R. Lupski** Baylor College of Medicine, Texas, USA
17:20 – 17:30	**Poster Awards by Rashid Al Ali**
17:30 – 17:35	**Closing Remarks by Donald Love**