Functional Genomics 2018

Big Data to Clinic

sidra.org

سدرة للطب
Sidra Medicine

The 4th Functional Genomics 2018

Big Data to Clinic

8 to 10 December 2018
Qatar National Convention Center
Doha, Qatar

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.

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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.

Message from the Chair

Dear Colleagues,

On behalf of Sidra Medicine, it is our pleasure to welcome you to Sidra Medicine's Fourth Functional Genomics. The theme of this year's symposium is "Big Data To Clinic". The field of genomics medicine has moved very rapidly from having the sequence of the genomes of thousands of individuals to clinical applications in prevention, diagnosis and therapy.

Qatar National Vision for 2030 is to build academic knowledge and health care through personalized medicine. Many actions have been initiated in the last five years and they are now materialized in training programs in genomics, precision medicine and genetic counselling. A systematic characterization of the population of Qatar is being performed through deep clinical evaluation and biobanking of samples by Qatar BioBank (QBB) that are being evaluated using multi-omic approaches by Sidra Medicine and other partners.

The Qatar Genome Program (QGP) has accomplished the pilot phase with high success, achieving the primary goals of i) obtaining the whole genome sequence data of 10,000 participants, and ii) performing the bioinformatics analysis of the sequence of these genomes. It has been announced by Her Highness Sheika Moza bint Nasser that the next phase will be Qatar Precision Medicine with a target on clinical cohorts, functional evaluation of mutations of the Qatari population, and on translation of genomic knowledge to the clinic.

This year's symposium will focus on the many aspects of this field. Our scientific committee has designed a program that encompasses the message of the genes through to the complex analysis of genomes; the latter including that of the Qatari population. The program highlights the modeling of disease in cells, zebrafish and mice, understanding disease prevention by sequencing patients at different time points, and altering the trajectory of genetic disease using cellular and genetic approaches.

Qatar is building the next generation of scientists that will base the future of the country on knowledge and technology. The symposium will be an excellent forum to discuss about the challenges that the field faces and should enhance collaborations between our scientists and the international experts that we have brought to Qatar. Research in genomics and precision medicine cannot be done in isolation. Qatar is ready to participate in international consortia and global efforts and challenges. The particularities of the population of Qatar with recent founder populations, the large families and the high rate of consanguinity should bring an enormous value to understand the major disorders that are affecting the population in the region and the world.

The success of the symposium is the result of our excellent scientific and organizing committees and the technical personnel and Sidra Medicine volunteer staff working diligently behind the scenes. Our gratitude to Sidra Medicine for being the main sponsor of the symposium but also to all the other organizations that have contributed to make it possible.

Finally, we would like to thank all participants and ask everyone to engage in open discussions with the speakers to make of the symposium a learning event for everyone. We hope you will enjoy the "Big Data To Clinic" Symposium and your time in Qatar.

Respectfully Yours,

Dr. Rashid Al Ali
Chair, Functional Genomics
Deputy Chief Research Officer
Chair, Bioinformatics, Research Department
Sidra Medicine

Professor Xavier Estivill
Chair, Functional Genomics
Chair, Genetics, Research Department
Sidra Medicine

Overview

Sidra Medicine is proud to host its fourth annual Functional Genomics from 8-10 December 2018 titled “Big Data to Clinic”. This year’s symposium will provide an opportunity to discuss cutting edge advances in genetics, genomics and “big data” in the field of medicine.

Understanding the variation of the human genome as well as its analysis and the evaluation of functional consequences of genetic variability requires extensive research, investment and collaboration. This needs the convergence of different communities of scientists to address the complexity of human disease and genome biology. What better platform to address this than at our very own Functional Genomics this year?

The “Big Data to Clinic” Functional Genomics will bring together a cross-disciplinary set of speakers who can address all aspects of genome variability and function, with a specific focus on the genetics of disorders affecting children.

The symposium will assemble a unique and highly interdisciplinary international community to discuss how best to use big genetic and clinical datasets to provide novel insights into the biology of disease. The symposium will also focus on various scientific challenges including implementation, scalable data infrastructures, the analyses required for gene discovery, as well as how to translate genomics findings for clinical care and the development of novel therapeutics.

Conference Objectives

1. Provide the latest scientific information on the collection, analysis and use of big data in the field of genomics and precision medicine;

2. Determine the steps involved in evaluating the functional consequences of genetic variants in the genome with respect to clinical phenotypes;

3. Explore the translation of the genomic knowledge for the prevention, diagnosis and treatment of genetic rare and common disorders that affect children.

Scientific Sessions

- The message of the genes
- Modeling human disease
- Reproduction and infertility
- Genome sequencing in action
- Cellular and organ models of disease
- Diagnosis of the future child
- Qatar genome and medical knowledge
- Treating genetic diseases
- Patient genome sequencing.

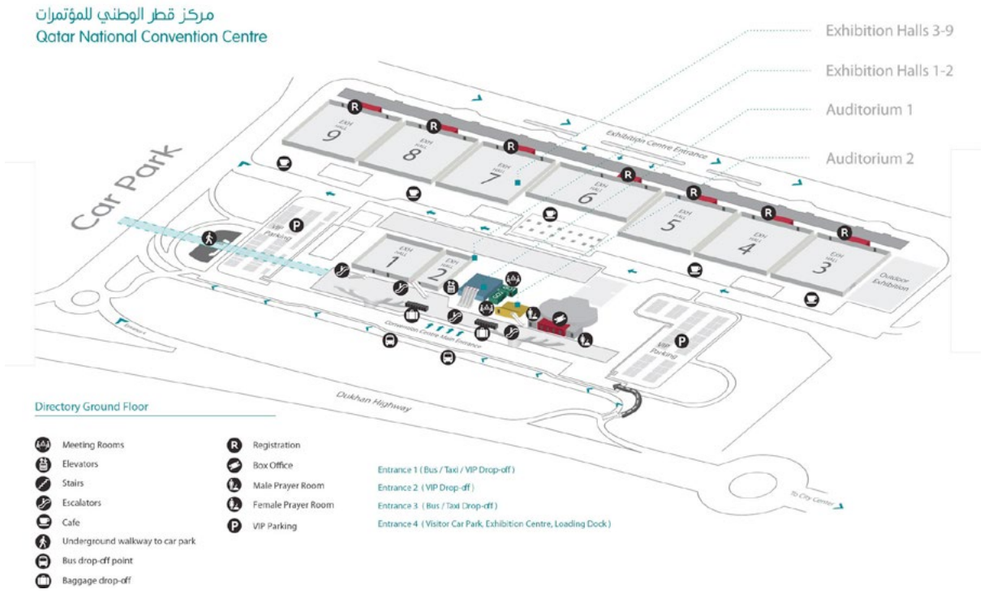
The Scientific Organizing Committee

Dr. Donald Love, Chief of Genetics Pathology
Dr. Bernice Lo, Investigator
Dr. Jithesh Puthen, Investigator
Dr. Rashid Al-Ali, Investigator
Dr. Xavier Estivill, Investigator
Dr. Tomoshige Kino, Investigator
Dr. Luis Saraiva, Investigator
Dr. Chiara Cugno, MD
Ms. Maribeth Caritan, Occupational Health Nurse
Ms. Asma Al Naama, Associate-Research
Mr. Mostafa Abdelhafız, Supervisor, Pharmacy Drug Supply

The Supporting Organizing Committee

Aisha Al Zaman, Specialist - Community Relations
Nelly EL Mistekawy, CRO Office Manager
Maria Theresa Brunsman, Executive Assistant
Rana Hamada, Executive Assistant
Maricris Salud, Executive Assistant
Nevin Amin, Sr. Admin Assistant
Kimberly Nieto, Sr. Admin Assistant

Venue



EDUCATIONAL OBJECTIVES

- 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.

TARGET AUDIENCE

The Functional Genomics Symposium is directed towards physicians, nurses, pharmacists, healthcare policy makers, scientists, academic researchers and students involved in the field of biology, genomics, genomic medicine, genetics, cancer and other chronic diseases.

PROGRAM SCHEDULE

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

07:00 – 07:40 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 Series
Xavier Estivill
Co- Chair of the Functional Genomics 2018

SESSION 1: THE MESSAGE OF GENES

MODERATORS: NICOLE SORANZO AND JITHESH PUTHEN

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

08:15 – 08:50 "From Genomics To Therapeutics: Uncovering And Manipulating The Genetic Circuitry of Human"
Manolis Kellis
MIT Computer Science, CSAIL, Broad Institute, Massachussetts, USA

08:50 – 09:25 "Tohoku Medical Megabank Project- A National Challenge to Realize Personalized Medicine"
Masayuki Yamamoto
Tohoku Medical Megabank Organization, Sendai, Japan

09:25 – 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”
Jithesh Puthen
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

08:15 – 08:50 “The Use of Functional Genomics to Accelerate Drug Discovery”
Nicholas Katsanis
Duke University, North Carolina, USA

08:50 – 09:25 “Long noncoding RNAs: A Potential Gold Mine of New Disease Genes”
Rory Johnson
University of Bern, Bern, Switzerland

09:25 – 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 – 08:15 **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

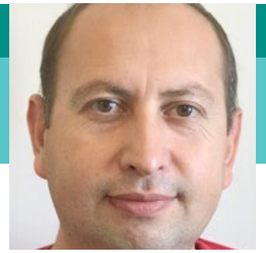
16:30 – 16:45 Mutation in the GTPase GIMAP6 Leading to Reduced Autophagy in a Severely Immune Deficient Patient"
Brittany Chao (Travel Award)
Oxford University, Oxford, UK

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**

SPEAKERS' BIOGRAPHIES



AHMET OZEN

**Prof. of Pediatrics, Chief Division of Allergy and Immunology,
Marmara University School of Medicine
Director of Istanbul Jeffrey Modell Foundation Diagnostic Center For Primary Immune Deficiencies, Istanbul, Turkey**

Dr. Ahmet Ozen is a Professor of Pediatrics and the acting division chief of Allergy and Immunology in Marmara University, Istanbul, Turkey since 2016. His program is focused on training residents in pediatrics and clinical fellows in Pediatric Allergy and Immunology. His institution is a referral center in Turkey for the diagnosis and treatment of patients with various immunological disorders, including childhood allergies and primary immune deficiencies (PID). He worked as a visiting fellow at Laboratory of Immunology, NIAID, at the National Institutes of Health, USA between 2014 and 2016 to receive advanced training in the field of genomics research of immunological disorders.

ANNE FERGUSON-SMITH

Arthur Balfour Professor of Genetics and Head of the Department of Genetics at the University of Cambridge

Dr. Anne Ferguson-Smith is a mammalian developmental geneticist and epigeneticist. An expert on genomic imprinting, her team studies the epigenetic control of genome function with particular emphasis on epigenetic inheritance. Her group is made up of both experimental and computational scientists and current research focuses on three themes: (i) Stem cells and the epigenetic programme, (ii) Functional genomics and epigenomics, and (iii) the interaction between the environment and development, health & disease. She was elected to EMBO in 2006, to the UK Academy of Medical Sciences in 2012 and became a Fellow of the Royal Society in 2017.

ANTONIO SCIALDONE

Junior Group Leader, Institute of Epigenetics and Stem Cells, Helmholtz Zentrum München
Munich, Germany

Dr. Antonio Scialdone is a Junior Group Leader at the Helmholtz Zentrum München (HMGU). He uses a multidisciplinary approach to understand the fundamental biological processes behind cellular decision making. Recently developed experimental techniques have made it possible to generate a vast amount of large-scale biological data at the single-cell level. By using state-of-the-art and newly developed computational methods, we combine information obtained from the analysis of these data with the insights offered by physical models that can guide interpretation. He works in collaboration with several experimental groups, focusing on biological questions concerning different systems, like mouse embryos and mouse olfaction.

CHRISTOF VON KALLE

Chief Research Officer
Sidra Medicine

Dr. Christof von Kalle joined Sidra in May 2018. In his role as the Chief Research Officer, he oversees, coordinates and directs all aspects of research conducted at the institution. His goal is to align the activities of the clinical and research branches of Sidra Medicine into one effective program aimed at developing diagnoses that improve the lives of patients, as well as make essential contributions to Qatar's healthcare and medical education systems.

As a physician scientist with a clinical background in hematology and oncology and over 150 acclaimed publications, Prof. Kalle is an internationally recognized research leader in stem-cells, mutation analysis and gene therapy.

Prof. Kalle joined Sidra Medicine from the National Center for Tumor Diseases (NCT) where he served as Director and chaired the NCT Board of Directors. At the NCT, Dr. Kalle coordinated overall activities, with the primary goal of facilitating excellence in translational and clinical research.

In addition to his role at Sidra Medicine, he retains his position as the Head of the Department of Translational Oncology, NCT and German Cancer Research (DKFZ) in Heidelberg, Germany.

CISCA WIJMENGA

Lodewijk Sandkuijl Professor of Human Genetics
University of Groningen and the University Medical Center Groningen, The Netherlands

Dr. Cisca Wijmenga moved to the University of Groningen in 2007 and now leads a research group of some 15-20 researchers working in the field of gut health using genomics technology to understand health and disease. Her work is focusing on the genetics of celiac disease, the role of the gut microbiome in health and disease and the development of gut-on-chip for more mechanistic research. Her research is interdisciplinary, encompassing the fields of genetics, molecular genetics, epidemiology, immunology, microbiology, computational biology and bioinformatics.

Over the past 10 years, her research has led to the identification of some 40 genetic risk factors that collectively explain 50% of the genetic variation (single nucleotide polymorphisms or SNPs) contributing to celiac disease, one of the most common diet-induced gastrointestinal diseases affecting some 1% of Western populations. Her group has strong bioinformatics expertise and uses this in systems approaches to model regulatory gene networks perturbed by celiac disease associated genetic variation or physiological stressors like infectious agents or dietary gluten. To this end she founded a functional genomics cohort (LIfeLines Deep) of ±1500 individuals including detailed phenotypes (~1500 clinical, lifestyle, dietary features) and multi-layered 'omics' data (genetic, transcriptomic, epigenetic, metabolite, microbiome).

DIEGO BOHORQUEZ

Cell and Molecular Biology Program
Duke University

Dr. Diego Bohorquez is a neurocientist and leader of Duke University's research laboratory on gut-brain biology. He was born in the Ecuadorian Amazon. His background is an amalgam of a B.S. in agriculture, a Ph.D. in Nutrition, and Postdoctoral training in the Neurosciences. At Duke University, Diego is the head of a research team teasing apart neural circuits connecting gut sensing with behaviors, like mood and appetite. Diego discovered a direct connection between nerves and enteroendocrine cells – a type of sensory cell dispersed throughout the gastrointestinal epithelium (the gut's internal lining). This discovery has opened a new field in sensory neurobiology – the field of gut-brain sensory transduction.

DONALD LOVE

Division Chief - Pathology Genetics
Sidra Medicine

Dr. Donald Love focuses on pathology genetics at Sidra Medicine. He undertook his PhD in the Biochemistry Department of The University of Adelaide studying extra-cellular protease and amylase synthesis by Bacillus amyloliquefaciens. The aim of this work was to gain insight into the molecular biology of prokaryote extracellular enzyme synthesis and secretion.

Dr Love moved to Auckland City Hospital in 2007 to become its Director of Diagnostic Genetics where his principal scientific role involved the development of new approaches to molecular-based diagnostics using innovative technology. These approaches concerned the implementation of bioinformatic approaches for the rapid design and evaluation of primers for the amplification of human disease-causing genes; the implementation of array-based molecular karyotyping; the implementation of custom array-based analysis of multiple genes to screen for deletion/duplication mutations; the implementation of massively parallel sequencing (MPS) strategies to replace conventional capillary-based Sanger-type sequencing to screen for mutations in the BRCA1 and BRCA2 genes; and the accreditation of his laboratory for Pathology Training in the discipline of Genetic Pathology- Medical Genomics (Royal College of Pathologists of Australasia). Dr Love was granted an Adjunct Professorship at AUT University in 2016.

ELIZABETH PHIMISTER

Deputy Editor
New England Journal of Medicine

Elizabeth (Bette) Phimister is a Deputy Editor at the New England Journal of Medicine where she evaluates research manuscripts describing genetic, genomic and ophthalmologic research. She also solicits and prepares commentaries on the clinical implications of basic research. She joined the Journal in 2002.

Before joining the Journal, Bette served as Editor-in-Chief of Nature Genetics, the world's leading genetics journal. She trained in molecular oncology at the Imperial Cancer Research Fund (now Cancer Research, U.K.) and was a fellow in molecular diabetology at Cambridge University until 1996, when she joined Nature Genetics. Her formative years were spent in Philadelphia, Pennsylvania, and the United Kingdom.

In addition to evaluating research manuscripts, Bette writes editorials and commentaries on translational research published in the Journal and elsewhere.

EULALIA MARTI PUIG

Associate Professor
University of Barcelona

Dr. Eulalia Marti Puig leads the Functional Genomics of Neurodegenerative Disorders group at the Department of Biomedical Sciences of the University of Barcelona and is Unit leader of the Centro de Investigación Biomédica en Red de Epidemiología y Salud Pública.

She has over 20 years of research experience in neurobiology, focusing on the molecular bases of neuronal dysfunction in pathological conditions. Her research is currently focused in the identification of non-coding RNA (ncRNA) mechanisms contributing to the onset and progression of age-related neurodegenerative disorders. To accomplish this her group has developed tools to identify ncRNA signatures from sequencing data and implemented molecular and cell biology approaches for functional screenings. The final purpose is to uncover ncRNA-gene expression networks underlying neuropathogenic processes with the aim to understand disease mechanisms and identifying pathways for therapeutic intervention.

EVAN EICHLER

Professor & Investigator, Department of Genome Sciences, University of Washington School of Medicine & Howard Hughes Medical Institute, USA

Evan Eichler, Ph.D., is a Professor and Howard Hughes Medical Institute Investigator (appointed 2005) in the Department of Genome Sciences, University of Washington School of Medicine in Seattle, Washington. Dr. Eichler joined the University of Washington in 2004 as an Associate Professor with tenure and then Professor in 2008. He also became an Associate Member of the New York Genome Center (NYGC) in 2015.

Dr. Eichler's research group provided the first genome-wide view of segmental duplications within human and other primate genomes and he is a leader in an effort to identify and sequence normal and disease-causing structural variation in the human genome. The long-term goal of his research is to understand the evolution and mechanisms of recent gene duplication and its relationship to copy number variation and human disease.

HENNING HORN

Assistant Professor
Hamad Bin Khalifa University

Dr. Henning Horn received his Ph.D. in Cell and Molecular Biology in 2003 from the University of Cincinnati. He then went on to study the role of ribosomal stress in activating the tumor suppressor p53 pathway. In 2008 he moved his research into nuclear envelope proteins and has analyzed several mouse models of these proteins (KASH5 and Nesprin 4). Using these models, he discovered roles for nuclear envelope proteins in infertility and hearing loss. Dr. Horn has been an Assistant Professor at HBKU since 2014, where his lab focuses on how cellular mechanics impacts cancer invasion, hearing loss, and infertility.

HUGO BELLEN

Professor in the Departments of Molecular and Human Genetics and Neuroscience at Baylor College of Medicine (BCM)

Dr. Hugo Bellen started his independent career as a Howard Hughes Medical Institute Investigator at BCM in 1989. He studies human disease genes and has utilized the fly to validate dozens of novel genes associated with neurological disease. In collaboration with human geneticists, he has elucidated the pathogenic mechanisms underlying neurodevelopmental and neurodegenerative diseases using the fly as a model organism. Dr. Bellen was chosen to lead the Model Organism Screening Center (MOSC) of the Undiagnosed Diseases Network to assess human variants. In the past few years the MOSC has contributed to the discovery of numerous previously unknown diseases. Dr. Bellen has also made probing discoveries related to Friedreich ataxia, Amyotrophic lateral sclerosis, Parkinson disease, Alzheimer disease, and others. In doing so, he has cemented the status of Drosophila as a model system for human disease gene validation, understanding of pathogenic mechanisms, and as a tool for translational therapeutics.

JAMES R. LUPSKI

Cullen Professor of Molecular and Human Genetics and Professor of Pediatrics, Baylor College of Medicine, Houston, Texas 77030 USA

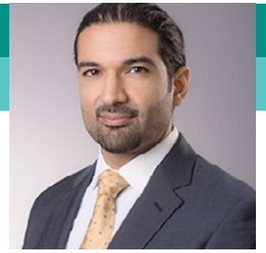
Dr. James R. Lupski is Cullen Professor of Molecular and Human Genetics and Professor of Pediatrics and is ABMG certified in Clinical Genetics and Clinical Molecular Genetics (1993-2022). He received his initial scientific training at the Cold Spring Harbor Laboratory as an Undergraduate Research Participant (URP; summers of 1978 &'79) and at New York University obtaining his undergraduate degree in chemistry and biology (1979) and completing the MD/PhD program in 1985. In 1986 he moved to Houston, Texas for clinical training in pediatrics (1986-1989) and medical genetics (1989-1992) and then established his own laboratory at Baylor College of Medicine where he remains, and as of 1995, as the Cullen Professor.

His research studies focus on trying to understand mutational mechanisms and linking specific mutations and genes to human disease. Dr. Lupski's studies of neurologic and ophthalmic Mendelian diseases have advanced current understanding of: i) gene dosage in trait manifestation, ii) structural variation (SV) mutagenesis, iii) parental mosaicism in transmission genetics and recurrence risk, iv) mutational burden including triallelic inheritance, and v) the importance of recessive carrier states in susceptibility to complex traits.

DAME KAY DAVIES

Professor of Anatomy
MDUK Oxford Neuromuscular Centre

Professor Dame Kay Davies is the Dr Lee's Professor of Anatomy and a co-Director of the MDUK Oxford Neuromuscular Centre. She is also Associate Head, Development, Impact and Equality, in the Medical Sciences Division at the University of Oxford. Her research interests cover the molecular analysis of neuromuscular and neurological disease, particular Duchenne muscular dystrophy (DMD). She has an active interest in the ethical implications of genetics research and the public understanding of science. She was Director of the MRC Functional Genomics Unit from 1999-2017. She has considerable experience of biotechnology companies as a conduit for translating the results of experimental science into new therapeutics and diagnostics. She is a co-founder of Summit Therapeutics and Oxstem. She has published more than 400 papers and won numerous awards for her work. She is a founding editor of "Human Molecular Genetics" and a founding fellow of the UK Academy of Medical Sciences. She is a Fellow of the Royal Society and a Member of the European Molecular Biology Organization (EMBO). She appointed a Governor of the Wellcome Trust in 2008 and was Deputy Chairman from 2013-17. She was made Dame Commander of the British Empire for services to science in 2008.

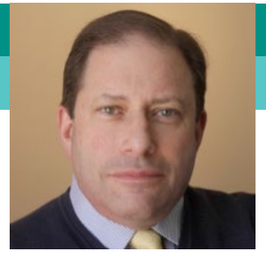


KHALID FAKHRO

Investigator - Human Disease Genetics
Sidra Medicine

Dr. Khalid Fakhro is Principal Investigator at Sidra Medical and Research Center. He obtained his Bachelor's degree with honors in Cell Biology and Molecular Genetics from the University of Chicago where his research focused on developing a doxycycline-inducible mouse model of Parkinson's disease. He subsequently pursued his PhD in Human Genetics at Yale University, where he was part of a highly selective Howard Hughes

Medical Scholars program focused on studying the basic molecular underpinnings of human disease. During his PhD training, Dr. Fakhro gained first-hand experience with multiple high-throughput data generating platforms including a range of high-density genotyping microarrays and next-generation sequencing platforms, as well the use of multiple model organisms to study human disease. Currently, his work focuses on employing new genomics technologies to discover responsible genes in patients with Mendelian disease and previously undescribed congenital syndromes. Dr. Fakhro holds an appointment as an Assistant Professor at Weill Cornell Medical College-Qatar in the Department of Genetic Medicine, where he teaches, maintains active research collaborations and mentors students through the Nationals Program.



LEE P. SHULMAN

Chief of the Division of Clinical Genetics
Feinberg School of Medicine of Northwestern University – Illinois, USA

Lee P. Shulman MD is the Anna Ross Lapham Professor in Obstetrics and Gynecology and Chief of the Division of Clinical Genetics at the Feinberg School of Medicine at Northwestern University in Chicago, Illinois. He also serves as the Co-Director of the Cancer Genetics Program of the Robert H. Lurie Comprehensive Cancer Center of Northwestern University, the Director of the Northwestern Ovarian Cancer Early Detection and Prevention Program and the Medical Director of Insight Medical Genetics and Reproductive Genetics Innovations. Dr. Shulman is also an Adjunct Professor in the Department of Medicinal Chemistry and Pharmacognosy at the University of Illinois at Chicago College of Pharmacy. He is a Fellow of the American College of Obstetricians and Gynecologists and a Founding Fellow of the American College of Medical Genetics.

LEILA JAMAL

Certified Genetic Counselor
National Institute of Allergy and Infectious Diseases (NIAID)

Dr. Leila Jamal received her BA in Philosophy, Politics, and Economics from Oxford University in 2002. She earned her ScM in genetic counseling from Johns Hopkins University and the National Institutes of Health in 2012 and her PhD in bioethics and health policy from the Johns Hopkins Bloomberg School of Public Health in January of 2017. She is also a practicing cancer genetic counselor at the University of Maryland. Leila's research interests include rare disease research ethics and the regulation of new technologies in healthcare and public health. Her dissertation focused on the roles of patient advocates in rare disease research policy. During her fellowship, Leila will develop an ethical framework for returning genetic results to patients and their relatives who are enrolled in the Baylor-Johns Hopkins Centers for Mendelian Genomics, an undiagnosed disease research project. She is also interested in developing novel approaches for sharing genetic information within families while respecting individual privacy.

LUIS PEREZ-JURADO

Professor
Pompeu Fabra University Barcelona

Luis A. Pérez-Jurado, MD, PhD, is Clinical Geneticist in the Women and Children Health Network, Senior Research Fellow in the South Australian Health and Medical Research Institute (SAHMRI) and Clinical Professor in the University of Adelaide, Australia (since 2017). He has also joint appointments as Professor of Genetics at Universitat Pompeu Fabra (UPF) (since 2010), and Senior Consultant and Group leader in the Neuroscience Program of the Hospital del Mar Research Institute, in Barcelona, Spain. He is a Steering Committee Member and Coordinator of Formation in the Spanish Network Centre for Biomedical Research on Rare Diseases (CIBERER) (since 2007). His research group has done significant contributions to the field of genetics of childhood onset neurodevelopmental disorders. The main goal of his research is to facilitate and drive translational research into the prevention of childhood disability integrating clinical practice with genetics and genomic tools. He got his Medical Degree (1983) and PhD (extraordinary award, 1992) in the Autonomous University of Madrid, and holds medical specialties in Family Practice (Granada, Spain), Paediatrics (Madrid, Spain), Clinical Genetics and Clinical Molecular Genetics (Stanford University, USA).

MANOLIS KELLIS

Professor
Broad Institute, USA

Manolis Kellis is a Professor of Computer Science at MIT, an Institute Member of the Broad Insti-tute of MIT and Harvard, a member of the Computer Science and Artificial Intelligence Lab at MIT, and head of the MIT Computational Biology Group (compbio.mit.edu). His research spans an unu-sually broad spectrum of areas, including disease genetics, epigenomics, gene circuitry, non-coding RNAs, comparative genomics, and phylogenetics. He has helped direct several large-scale ge-nomics projects, including the Roadmap Epigenomics project, the ENCODE project, the Roadmap Epigenomics Project, the Genotype Tissue-Expression (GTEx) project, and comparative genomics projects in mammals, flies, and yeast. He received the US Presidential Early Career Award in Sci-ence and Engineering (PECASE) by US President Barack Obama, the NSF CAREER award, the Alfred P. Sloan Fellowship, the Technology Review TR35 recognition, the AIT Niki Award, and the Sprowls award for the best Ph.D. thesis in computer science at MIT. He has authored over 180 journal publications, which have been cited more than 68,000 times. He lived in Greece and France before moving to the US, and he studied and conducted research at MIT, the Xerox Palo Alto Re-search Center, and the Cold Spring Harbor Lab.

MARGARET ELIZABETH ROSS

Professor of Neurology
Weill Cornell Medical College

Dr. M. Elizabeth Ross received her M.D. from Cornell University Medical College, Ph.D. from Cornell University Graduate School of Medical Sciences, and her training in Neurology at Massachusetts General Hospital, Harvard Medical School, and molecular genetics at Harvard and Rockefeller University. She directs the Center for Neurogenetics in the Brain and Mind Research Institute, Weill Cornell Medicine, which supports research into the genetic causes of neurological disorders in children and adults. The Center has both basic science and clinical arms, evaluating patients with neurological disorders of monogenic or complex genetic origin and operates the biobank for the neurological community at Weill Cornell. Neuroscientist faculty members of the Center investigate the mechanisms underlying pathogenesis of these conditions. Her own research group, the Laboratory of Neurogenetics and Development, focuses on discovery of gene mutations associated with brain malformations and investigation of how these genes direct the construction of brain.

MARIAM ALMAADEED

Vice President for Research and Graduate Studies
Qatar University

She is leading the research and graduate studies strategies, programs and developments. Prior to that, professor Mariam worked as the director of the Center for Advanced Materials and the founder and Program coordinator of Materials Science and Technology program. She wrote more than 200 peer reviewed publications in international journals and conferences, book chapters, patents and edited four books. Professor Mariam received many grants and led several projects with a number of universities and institutions around the globe, and national and international companies. She has considerable experience and international recognition in the materials and nanotechnology field. She supervised many graduate theses at Qatar University and other international universities. Professor Mariam worked as a consultant and presented several workshops for a variety of organizations including industrial companies and ministries. She chaired several conferences, was invited as a keynote speaker to many international conferences and was a member of a number committees in the country and abroad. Professor Mariam has also worked as a reviewer for many scientific journals and conferences.

MASAYUKI YAMAMOTO

Professor
Tohoku Medical Megabank Organization

Masayuki Yamamoto graduated from Tohoku University School of Medicine and Graduate School of Medicine. In 1983, he obtained Doctor of Medical Sciences. In 1983-1986, Yamamoto was a postdoctoral fellow at Northwestern University with Professor Doug Engel, and cloned erythroid-type 5-aminllevulinate synthase (ALAS2) cDNA. In 1989, Yamamoto revisited the Engel laboratory and in collaboration identified the GATA family of transcription factors, one of the prototype transcription factor families regulating lineage commitment and cell differentiation. In 1991, Yamamoto returned to Japan and started analyses of the Gata1 and Gata2 genes. In 1995, Yamamoto became a Professor at University of Tsukuba and started a series of analyses on CNC and sMaf family of transcription factors. He identified KEAP1-NRF2 system regulating cellular response against electrophilic and oxidative stresses. Yamamoto was awarded with the Purple Ribbon (2012) medal and the Japan Academy Prize (2014). In 2012, he started Tohoku Medical Megabank Project for creative reconstruction of the Tohoku area from the devastating earthquake and tsunami occurred on March 11, 2011. The project aims to establish two types of genome cohorts and an integrated biobank.

MEHDI ADELI

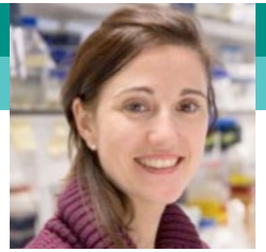
Senior Consultant, Pediatric Allergy and Immunology, Sidra Medicine
Assistant Professor, Weill Cornell Medical College –Qatar

Dr. Mehdi Adeli is an Assistant Professor in Weill Cornell Medical College in Qatar and a Senior Consultant in Pediatric Allergy and Immunology in Sidra Medicine. He is the Leader in formulating the Vision of Allergy and Immunology service and plan for Sidra Medicine and also the Leader in creating a multidisciplinary approach center for Eczema in Qatar.

His most important work experiences include: Establishing the Qatar National Neonatal Screening Program for Severe Combined Immunodeficiency and the Qatar National Registry for Primary Immunodeficiency Diseases, developing the Aeroallergens Mapping Project in Qatar for detection of allergens. He started the Allergy and Immunology Awareness Program (AIAP) in 2014 to improve patients care through patients education (Booklets, Leaflets, DVDs).

Dr. Adeli obtained his medical degree from Damascus University Medical School in 1993 and was certified with Arab Board in Pediatrics since 1997. He then joined Inova Fairfax Hospital for Children in Falls Church Virginia, USA

where he was certified with American Board in Pediatrics in 2006. After this he joined Duke University Medical Center, Durham NC where he completed his fellowship training program in Pediatrics Allergy and Immunology in 2008 and Allergy and Immunology advanced research training program in 2010 and certified with American Board of Allergy and Immunology in 2009. He re-joined Hamad Medical Corporation in 2010 before moving to Sidra Medicine in November 2017.



MERI HUCH

Group Leader
Wellcome/CRUK Gurdon Institute, University of Cambridge

After obtaining her PhD degree at the Center for Genomic Regulation in Barcelona, Spain, she moved to the Netherlands to join the laboratory of Professor Hans Clevers in order to redirect the focus of her research into Adult Stem Cell Biology. In her postdoctoral stay, she isolated adult mouse stomach and mouse and human liver cells and proved these can be expanded in culture, forming stomach and liver organoids in vitro. In 2014 Dr Meritxell Huch established her independent lab at the Gurdon Institute, University of Cambridge, where she works on elucidating the replicative potential of adult stem cells during tissue regeneration and disease. Research prizes and awards: 2018: The Women in Cell Biology Early Career Medal for 2018. The British Society for Cell Biology. “For pioneering research on stem cell, in particular the work on liver organoids”. 2016: The Hamdan Award for Medical Research Excellence. “For innovative methods and research that resulted in novel discoveries and inventions”. 2014: Wellcome-Beit Prize for outstanding scientists of the 2014 year as additional recognition to the success in obtaining Wellcome Trust Fellowship to become independent. 2014: NC3Rs International Prize. National Centre for the Replacement, Refinement & Reduction of Animals in Research, UK. “For growing ‘mini-livers’ in culture”. Since 2005, Dr Meritxell Huch has published a total of 46 publications.

NICHOLAS KATSANIS

Director, Center for Human Disease Modeling
Duke University

Nicholas Katsanis obtained his BSc from University College London in 1993 and his PhD from Imperial College London in 1997 where he first worked on the genetics of Down syndrome. He completed his postdoctoral work in the laboratory of Dr. Lupski at Baylor College of Medicine in Houston and transitioned his studies to Bardet-Biedl syndrome. In 2002, he established his independent research lab at the Institute of Genetic Medicine, John Hopkins University, where he led studies that unified several allied conditions under the ciliopathy umbrella. Dr. Katsanis demonstrated that Bardet-Biedl syndrome is caused by centrosomal/basal body dysfunction, establishing that dysfunction at the primary cilium can give rise to a large group of disorders with both clinical and genetic overlap (the ciliopathy model). In 2009, he moved to Duke University to establish the Center for Human Disease Modeling (CHDM), where he is the Director; this structure aims to facilitate collaboration across disciplines and to develop physiologically relevant tools to study variation found in human patient genomes.

NICOLE SORANZO

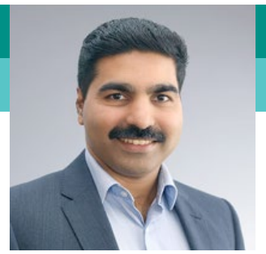
Professor
Wellcome Trust Sanger Institute (WTSI)

Prof. Soranzo is a human geneticist working in the field of human complex trait genetics. Her research focuses on the application of large-scale genomic analysis to unravel the spectrum of human genetic variation associated with cardiometabolic and immune diseases, and its interaction with non-genetic and environmental cues. Prof. Soranzo graduated in biological sciences at the University of Milano, Italy, with a dissertation on population and evolutionary genetics. She later obtained a PhD in genetics from the University of Dundee, and undertook post-doctoral training in human population and statistical genetics at University College London, conducting applied and methodological work in evolutionary genetics and association studies. In 2005 Prof. Soranzo joined the Pharmacogenomics Department at Johnson & Johnson Pharmaceutical Research and Development (Raritan, USA). In 2007 she joined the Wellcome Trust Sanger Institute, and since 2009 she has led her own team. In 2015 she was additionally appointed as Professor of Human Genetics at the School of Clinical Medicine of the University of Cambridge. Furthermore, Prof. Soranzo is a member of the Cambridge University Platelet Biology and Cardiovascular groups, the NIHR Blood and Transplant Research Unit in Donor Health and Genomics and the EU BLUEPRINT and EpiGeneSys projects. She serves in several steering committees and scientific advisory boards, and is on the editorial board for the European Journal of Human Genetics, Genome Medicine, Trends in Genetics, PLoS Biology and Molecular Biology and Evolution.

OMAR ALBAGHA

Principal Investigator/Professor of Genetics
QBRI/Hamad Bin Khalifa University

Dr. Albagha received a BSc degree (with distinction) in Medical Technology from the University of Jordan in 1996 and then he moved to the UK where he obtained an MSc in Medical Molecular Genetics from the University of Aberdeen in 1997. He was awarded a PhD scholarship to investigate genetic loci influencing susceptibility to osteoporosis at the University of Aberdeen. After completing his PhD degree in 2001, he worked as a postdoctoral researcher investigating the genetic determinants of osteoporosis until 2002 when he was appointed as an Arthritis Research UK lecturer (Assistant Professor) in Genetics of Bone disease at the Institute of Medical Sciences, University of Aberdeen, UK. In 2005 he moved to the University of Edinburgh as a Principal Investigator/Group leader at the Centre for Genomic and Experimental Medicine (CGEM). In 2014, he was appointed the Associate Director of the Paget's Association Centre of Excellence, Edinburgh. In early 2017, he joined Qatar Biomedical Research Institute as a Principal investigator and Professor of Genetics at Hamad Bin Khalifa University. His current research focuses on Genomic Medicine aiming at understanding the genetic determinants of complex disease with emphasis on Type 2 Diabetes, and Autism. He has active collaborations with Qatar Genome Project to investigate genetics of disease-related traits in Qatar. He has published in high impact journals (Nature Genetics, JAMA, Cell Reports, and JBMR,..) and his work has been featured in news outlets such as the BBC and other international media.



JITHESH PUTHEN

Group Leader in Translational Bioinformatics
Sidra Medicine

Dr Jithesh is a Group Leader in Translational Bioinformatics. He holds a Masters in Microbiology and Advanced Diploma in Bioinformatics as well as a PhD in Medical Bioinformatics from Queen's University Belfast, UK. He has over 20 years of research and development experience in a broad range of fields in biology, health and computing. He has experience in the development of computing infrastructure for the management and analysis of 'big data' in bioinformatics and also in the analysis of data from high throughput experiments such as microarrays and next generation sequencing in the context of biomedical research. Before joining Sidra, he was the Head of Bioinformatics for the Translational Molecular Diagnostic Centre at the University of Oxford and honorary scientist at the Wellcome Trust Centre for Human Genetics, Oxford.

ROB RICHARDS

Professor of Genetics
The University of Adelaide

Professor Richards joined The University of Adelaide as an Associate Professor in 2001 and was promoted to Professor in 2008. Prior to this he was Chief Molecular Geneticist at the Women's and Children's Hospital, Adelaide where he had commenced employment in 1989. This followed 4 years as a Senior Research Fellow at the Howard Florey Institute at the University of Melbourne (1985-1989) and this was preceded by 4 years as a Research Fellow at the Australian National University, Canberra (1981-1985). His laboratory's research focus is on the use of animal models to define the pathogenic pathways responsible for the clinical manifestations of genetic diseases, particularly neurodegenerative diseases. Most of these models are in Drosophila with some collaborative work in zebrafish. His experimental results and a growing list of research publications from other laboratories reveal that neurodegenerative diseases have genetic hallmarks of autoinflammatory disease that therefore provides a plausible common pathogenic mechanism.

RORY JOHNSON

Junior Group Leader, Assistant Professor
University Hospital of Bern, Switzerland

Since 2016, Prof Rory have been Junior Group leader with the Swiss National Centers for Competence in Research (NCCR) "RNA & Disease", and Assistant Professor at the Medical Faculty of the University of Bern.

He lead the Genomics of Long noncoding RNAs in Disease laboratory focussed on understanding the role of elusive long noncoding RNAs (lncRNAs) in common human diseases. We follow an interdisciplinary "wet/dry" strategy, including in-house genome-editing (CRISPR-Cas9) and transcriptome analysis tools, to screen for potential drug target lncRNAs.

SAID ISMAIL

Manager of the Qatar Genome Programme
QGP, Qatar

Dr. Said Ismail is the Manager of the Qatar Genome programme (QGP), which is one of the major national programs of its kind in the region. QGP is a population-based initiative designed around on a comprehensive strategy involving drafting regulations and policies; forging local research partnerships; establishing national genomic data networks; building local human capacity; and facilitating the integration of genomics into the healthcare system. Dr Ismail, a Ph.D. graduate from the University of Oxford, has work experience extending into various fields including education, research and consultancy. His research experience is in the field of molecular biology and genetics with focus on cancer genetics and the screening and characterisation of disease related mutations in the Arab population. Dr Ismail is also the first president and co-founder of the International Society on Aptamers, and is also an editorial board member in multiple international journals. As a research fellow at University College London (UCL), Dr. Ismail won the Said Foundation prize for young Arab researchers in the UK. As head of the Biochemistry department at the University of Jordan Medical School, Dr Ismail was awarded the Shoman award for Arab researchers in biomedical sciences for the year 2013.

SARAH ROBERTSON

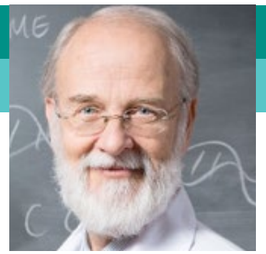
Director of the Robinson Research Institute
University of Adelaide

Professor Sarah Robertson is Director of the Robinson Research Institute, University of Adelaide. Her research focus is the immune response to conception and pregnancy, and consequences for reproductive success and offspring health. Defining fundamental biological pathways and developing novel interventions targeting immune pathways to tackle fertility and gestational disorders, is the goal of her work. She is funded by the NHMRC, ARC, CIHR and Gates Foundation, and has 170 peer-reviewed scientific journal papers and reviews. She is an elected Fellow of The Australian Academy of Science, the Australian Academy for Health and Medical Sciences, and Fellow of the Society for Reproductive Biology. She serves on the Editorial Boards of Endocrinology and the Journal of Clinical Investigation.

SOUHAILA AL KHODOR

Investigator - Division of Translational Medicine
Sidra Medical and Research Center, Qatar

Dr. Al Khodor is an Investigator in the Division of Translational Medicine at SIDRA Medical and Research center, Qatar, since January 2015. Dr. Al Khodor received her Bachelor's degree in Medical Lab. technology from the Faculty of Public Health at the Lebanese University in 2001. Soon after, she started her Masters degree in Microbiology and Immunology at the American University of Beirut while working as a Senior Microbiologist in charge at Hammoud University Medical Center in Lebanon (2002-2005). Dr. Al Khodor received her second Masters degree and her PhD in Microbiology and Immunology from the University of Louisville, Louisville, KY, USA (2005-2008). Her work focused on studying how Legionella pneumophila an intracellular pathogen, hijacks the host ubiquitination machinery. Before joining SIDRA, Dr. Al Khodor worked as a postdoctoral fellow in the Signaling Systems Unit, laboratory of Systems Biology, at NIAID NIH, USA. She used systems biology based approaches to study Burkholderia cenocepacia, a multidrug resistant intracellular pathogen that causes devastating infections in patients suffering from cystic fibrosis. At SIDRA, Dr. Al Khodor will pursue her interest in understanding the molecular mechanisms by which microbes signal through the cell, colonize the host, evade the immune response and cause infection. Moreover, using high throughput technologies she aims to study the role of the human microbiome in normal and pathological conditions and to develop models to study the host-microbiome interactions. She is interested in deciphering whether changes in the vaginal microbiome can predict woman's health and pregnancy complications and whether manipulating the gut microbiome composition can improve outcomes of Vitamin D treatment and kidney disease prognosis. Dr. Al Khodor has over 19 peer-reviewed publications, has membership of the American Society of Microbiology (since 2005) and is a member of the editorial board of the Frontiers in Microbiology (since 2015).



STYLIANOS ANTONARAKIS

Emeritus Professor
University of Geneva

Stylianos E. Antonarakis is currently Emeritus Professor (active) at the University of Geneva. He was previously Professor and Chairman of Genetic Medicine at the University of Geneva Medical School, and the founding director of iGE3 (institute of Genetics and Genomics of Geneva). He is a medical, molecular, human geneticist, physician-scientist, who studied extensively the relationship between genomic and phenotypic variation. He received his MD (1975) and DSc (1982) from the University of Athens Medical School, and after a specialization in Pediatrics in the University Hospital, Athens Greece, he moved to Baltimore, Maryland to the program of Medical Genetics at the Johns Hopkins University School of Medicine with

Haig H. Kazazian and Victor McKusick (1980-1983). He joined the faculty of the Johns Hopkins University in 1983 and rose to full professor of Pediatric Genetics, Biology and Medicine in 1990. In 1992 he moved to Geneva, Switzerland to chair Genetic Medicine in the University of Geneva. His research work includes the molecular bases of monogenic disorders and complex genetic disorders including the beta-thalassemias, hemophilias, and trisomy 21. His laboratory participated in the human genome sequence and functional analysis, particularly on chromosome 21. He is an international expert on disorders of chromosome 21, cloning of genes for genetic disorders, development of diagnostic tests, genome structure and function, studies of the genome variability, and conserved non-coding sequences in human DNA.

TOMOSHIGE KINO

Principal Investigator
Human Genetics

Dr. Kino's group investigate molecular actions of nuclear hormone receptors and their pathologic contribution to metabolic and reproductive diseases, such as diabetes mellitus, obesity, abnormal pregnancy and infertility. We do this by focusing on abundant non-protein-coding RNAs and gene regulatory elements, which are major drivers of human evolution and diversification, and thus, central components for determining individual susceptibility to these common diseases. My group retrieves valuable information from the Qatar Genome Program as well as performs bench experiments in model systems. We will develop new treatment/diagnostic means, ultimately contributing to the promotion of women's and children's health in Qatar.

XAVIER ESTIVILL

Acting Chief of Translational Medicine
Sidra Medical and Research Center, Qatar

My group is interested in evaluation of the genetics component of phenotypes that have clinical consequences for children and women. During the last years my group has examined the contribution of different types of genetic variation to human disease. These include non-coding RNAs, structural variants and epigenetic changes. We are using the knowledge of the Qatar Genome Program and the Qatar Biobank to dissect the genetic variability of the population and the phenotypic traits evaluated in this scientific endeavor. We are interested in cardio-vascular disorders, women infertility processes and psychiatric traits. We will develop translational genomics for woman and child's health. This will be achieved by developing studies in functional genomics that integrate large cohorts of patients with genome-centric evaluation of phenotypes. The group will help to make possible the concept of precision medicine in Qatar at prevention, diagnosis and treatment. Genomics translational activities will include the development of diagnostic approaches to prevent genetic disorders throughout carrier, neonatal and premarital screening. We will use model systems that include drosophila, zebrafish and induced pluripotent cells to model phenotypes affecting the population of Qatar. We will use high-throughput approaches to identify pharmaceutical compounds that will target every phenotype and patient under investigation.

MODERATOR'S BIOGRAPHIES

ASMA AL THANI

Founding Dean, College of Health Sciences
Qatar University

Dr Asma Al-Thani is a Professor of Virology at the Biomedical Science Department - College of Health Sciences. Dr Al-Thani obtained her Ph.D degree in (2005) from the University of London in the United Kingdom.

Since 2005, Dr Al-Thani has served as an Assistant Professor of Virology at Qatar University until 2011, when she was promoted to the position of Associate Professor and to Professor Position on 2016. Dr Al-Thani also fulfills roles as Adjunct Assistant Professor in the Department of Microbiology and Immunology at Weill Cornell Medical College in Qatar since 2011, as Vice Chairperson of the Qatar Biobank Board since 2010 at Qatar foundation, nominate to be Chair of National Genome Qatar Committee since December 2013, Director of Biomedical Research Center at Qatar University from Fall 2014 and Dean of College of Health Sciences on 2016.

BERNICE LO

Principal Investigator, Human Genetics (Immunogenetics)
Sidra Medicine

Bernice Lo is a Principal Investigator at Sidra Medicine in the Division of Translational Medicine. She joined Sidra in 2016.

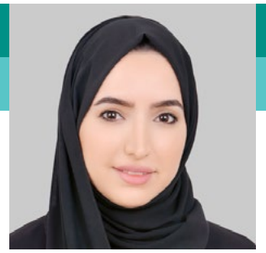
The research focus of Dr. Lo's laboratory is on understanding the molecular mechanisms of immune regulation and tolerance. Her approach includes identifying the genetic basis of disease in patients with immune dysregulation and autoimmunity with the ultimate goal of elucidating the molecular pathways involved and, therefore, revealing new therapeutic targets for disease treatment or novel drugs for immunomodulation. Dr. Lo's lab specializes in using cellular and biochemical techniques to uncover the role of newly discovered gene mutations in causing disease and in understanding the function of these genes in immune regulation. She has contributed to the discovery, diagnosis, and molecular understanding of multiple inherited autoimmune disorders.

EDWARD STUENKEL

Dean, College of Health and Life Sciences (CHLS)
Hamad Bin Khalifa University (HBKU)

Edward Stuenkel received a PhD based in Neuroscience from the University of Hawaii and conducted postdoctoral research at University of California, San Francisco and University of Michigan. Dr. Stuenkel advanced to Professor of Physiology within the Medical School at the University of Michigan. He is now Dean of the College of Health and Life Sciences at Hamad Bin Khalifa University.

At the University of Michigan Dr Stuenkel served as Director of the Neuroscience Doctoral Program and was director of NIH supported educational training programs in neuroscience. He was also Director of an International Graduate Education Initiative and key to leadership of a NSF supported program directed at Advancement of Neurotechnology.



KHOLOUD A SHAFAI

Senior Research Associate
Qatar Cardiovascular Research Institute

Kholoud Al-Shafai is a specialist in Genomic Research at Sidra Medicine. She obtained her Bachelor's degree with Honors in Human Genetics in 2009 from Leeds University in the United Kingdom where she was trained at the diagnostic laboratory of St. James hospital to genetically screen families with Congenital Stationary Night Blindness. After that, Kholoud joined Qatar Research Leadership Program (QRLP) at Qatar Foundation and was trained at the genetic laboratory of Weill Cornell Medical College-Qatar. In 2011, Kholoud earned her master's degree with distinction in Human Molecular Genetics from Imperial College London, UK where she studied DNA methylation of a-synuclein gene in Parkinson Disease progression.

LUIS SARAIVA

Principal Investigator
Sidra Medicine

Luis Saraiva is a Principal Investigator at Sidra Medicine in the Division of Translational Medicine, since October 2016. He was born in Portugal, where he completed a "Licenciatura" (BSc+MSc) in Biology at the University of Evora and Gulbenkian Institute of Science. He received his PhD in Genetics (summa cum laude) from the University of Cologne in Germany (with Sigrun Korsching). As a postdoc focused on the fields of molecular and behavioral neurosciences (with Linda Buck) and transcriptomics (with Darren Logan and John Marioni).

The main goal of his lab is to unravel molecular mechanisms underlying specific neural and metabolic processes and to understand their dysfunction in the brain and metabolic disorders. We have a special interest in the neurobiological basis of nutrition and obesity, and in understanding how genetic variation, environment, gender and age may impact these mechanisms. An additional interest of the lab includes the identification of gene variants underlying complex traits and neurological and neuroendocrine disorders. In his research he collaborate with clinicians and employ an integrative strategy combining a wide range of established techniques (molecular biology, anatomy, genetics, physiology and behavior) with omics technologies, resorting to both animal models and humans.

RICHARD O'KENNEDY

Vice President for Research
Development and Innovation (RDI)

Richard O'Kennedy, B.Sc., Ph.D., C. Biol., FRSB., F.I.Biol.I., Dip. F.S., Dip. C.S., Dip. Computing, MRIA, is Vice-President for Research, Development and Innovation at the Qatar Foundation and Vice-President for Research at Hamad Bin Khalifa University, Doha, Qatar, previously he was Professor and Scientific Director of the Biomedical Diagnostics Institute at Dublin City University (DCU). He is past President of the London International Youth Science Forum, Past President of the Institute of Biology of Ireland, past Chairman/ Founder of the Centre for Talented Youth (CTY) in Ireland and represented Ireland and the Royal Irish Academy (RIA) on the International Union of Biochemistry and Molecular Biology (IUBMB).

STEPHAN LORENZ

Director, Clinical Genomics Laboratory
Sidra Medicine

Dr. Stephan Lorenz graduated in Biochemistry at the University of Leipzig where he investigated the role of GPCR kinases in the regulation of GPCR activity. He then joined the laboratory of Prof Ralf Paschke in Leipzig for his PhD, studying calcium-binding proteins and their role in benign thyroid tumours. After his first PostDoc, he joined the Single Cell Centre at the Wellcome Sanger Institute in Cambridge in 2013 to develop novel single-cell whole-genome amplification methods in the group of Thierry Voet. A year on, he started to build and manage one of the first core facilities focussed on single cell genome and transcriptome sequencing. He and his team developed methods to rapidly process thousands of cells per experiment using robotics to achieve high throughput and high-end automation to minimize assay volumes and cost. In 2018, he joined Sidra Medical and Research as Director of the Clinical Genomics Core, where he is aiming to use his background in automation and miniaturization to enable more cost-effective sequencing solutions, thus supporting important initiatives like the Qatar Genome Project, but also enabling the use of sequencing technologies in a clinical setting.

ABSTRACT PRESENTERS' BIOGRAPHIES

ALICE ABDEL ALEEM

Principal Investigator of the Neurogenetics Laboratory
Weill Cornell Medicine Qatar, WCM-Q

Dr. Alice Abdel Aleem received her Medical Degree from Faculty of Medicine-Cairo University-Egypt and her Doctoral specialty training in Molecular Human Genetics from humangenetik Institute, Medical School Hannover-Hannover-Germany. Her training in clinical genetics and dysmorphology was received at the Clinical Genetics Department-Human Genetics Division- National Research Centre-Cairo-Egypt. She had her Postdoc training at several Genetic institutes involving Marburg Genetic Institute- Marburg University-Germany, Center Neuroscience California University-San Diego-USA, and Centre of Regenerative Bioscience, Georgia University-Athens-USA

Currently she is an Assistant professor at the Neurology Department - WCM-NY and Assistant Professor of Neuroscience at the Brain and Mind Research Institute –NY. She is also a a certified clinical Geneticist and a Consultant Geneticist at the Clinical and Metabolic Genetics- Hamad Medical Corporation.

ANDREA GUENNOUN

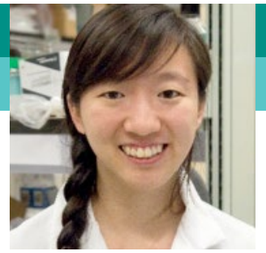
Staff Scientist, System Biology and Immunology Unit
Sidra Medicine

Andrea joined the Sidra Medicine Research department as a Staff Scientist in 2015. She leads three clinical research projects on primary immunodeficiencies in which she studies genetic defects in NFkB pathways in particular and more generally, susceptibility to severe infections in children.

Andrea holds a PhD from the University of Wuerzburg in Germany which was carried out in collaboration with the National Institutes of Health, Bethesda, US and Genelux Corp., San Diego, US in 2010. For her PhD thesis, she focused her research on an oncolytic Vaccinia Virus model as a therapeutic and diagnostic approach for immunologic tumor therapy which is now being tested in several clinical phase I/II trials.

Andrea then joined the Hematology & Oncology department at the University Hospital in Wuerzburg, Germany as a Postdoc and studied immune responses against tumor-associated antigens in the course of pregnancy as a model for allogeneic hematopoietic stem cell transplantations.

For the following three years, she was working in Weill Cornell Medicine-Qatar as a PostDoc and investigated genomics and epidemiology of metabolic disorders in Qatar and the interaction of white and brown adipose tissue with infiltrating immune cells.



BRITTANY CHAO

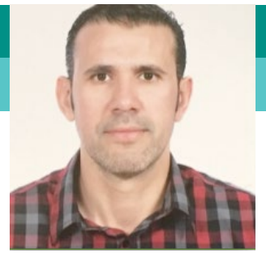
Postdoctoral Fellow
University of Oxford

Brittany Chao graduated from Cornell University in 2012 with a B.A. in Biological Sciences and a concentration in Microbiology. As a Gates Cambridge scholar, she then studied at the University of Cambridge and conducted research in the laboratory of Dr. Philip Stevenson. There, she studied the pathology of gamma herpesviruses using a murine model and received her Master's in 2013. Afterwards, she was accepted into the NIH OxCam program for her Ph.D., where she works with Profs Katja Simon and Andrew McMichael at the University of Oxford and with Dr. Michael Lenardo at the NIH. Her graduate research focuses on identifying novel genes associated with primary immunodeficiencies and understanding the importance of these genes in human immunology. Recently, Brittany has been studying a family of proteins called the GTPases of immunity-associated proteins (GIMAP), and has identified a role for GIMAP6 in lymphocyte autophagy.

GAURAV THAREJA

Senior Research Specialist, Bioinformatician Geneticist
Weill Cornell Medical College in Qatar

Gaurav Thareja is a Sr. Bioinformatics Data Specialist at Weill Cornell Medicine – Qatar. He is working on ways by which integrative omics can provide holistic view of human traits. Currently, he is looking at the genetics and epigenetics mechanisms impacting RNA expression and their effects on human traits. Previous to joining Weill Cornell Medicine – Qatar, his work focused on population genomics of Kuwaiti population.



GHEYATH KHALED NASRALLAH

Associate Professor, Biomedical Science
Qatar University

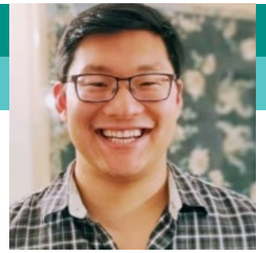
He is currently works as an Associate professor at the Biomedical Science, Qatar University. Gheyath research interests span two different fields; infection and immunity and zebrafish research

KARINA CERVANTES

Student- FG 2018 Travel Awardee
Genetics and Molecular Biology Department

Karina was born on October 10th, 1992 in San Juan del Río Querétaro México. She obtained the bachelor's degree in Chemistry Pharmacobiology from the Universidad Autónoma de Nayarit in 2016. She completed her Master at the Department of Genetics and Molecular Biology of CINVESTAV-IPN under the direction of Dr. Jaime García Mena in 2018, Studying how breast milk microbiota is related to the bacterial entero-mammary route and its effects on the infant gut microbiota.

She is currently a PhD student at CINVESTAV-IPN with the current study on Immunology and microbiology of the breast milk.



KEVIN ZHU

Predoctoral Fellow, Duke University
Department of Molecular Genetics and Microbiology

Kevin Zhu was born in Canada and completed his undergraduate degree at UC Berkeley in Microbiology while performing research with Dr. Patricia Pesavento at the UC Davis School of Veterinary Medicine. In the Pesavento lab, Kevin applied molecular biology and pathology to discover and characterize a diverse array of viral pathogens affecting household pets, farm animals, and wildlife. After graduation Kevin joined Radiant Genomics, a metagenomics driven natural product discovery startup, where he carried out the high-throughput construction of a genome-wide, gene-specific knockout library for the pathogenic yeast, Cryptococcus neoformans. After joining Duke's department of Molecular Genetics and Microbiology in 2015, Kevin worked with Dr. Lawrence David to setup and optimize microbiome meta-transcriptomics on a High-Performance Computing Cluster before joining the lab of Dr. Hiroaki Matsunami. Currently Kevin is a Ruth L. Kirschstein predoctoral fellow and in the Matsunami Lab his thesis work combines targeted transcriptomics with spatial samples to determine the target sites of olfactory neuron projections in wild-type mice, a long-standing and fundamental problem in olfaction.

NAYRA AL THANI

Research Specialist
Weill Cornell Medicine – Qatar

Nayra M. Al-Thani is Research Specialist in the Functional Genomics Laboratory at Weill Cornell Medicine - Qatar (WCM-Q). She received her B.S. in Biology and a Minor in Chemistry from Qatar University in 2015. Prior to joining WCM-Q as Research Specialist, she enrolled summer training program and gained experience in Type 2 Diabetes research in Life Science Research at Anti-Doping Laboratory Qatar. She entered WCM-Q as an Intern under the Biomedical Research Training Program for National. This work resulted in the publication of "Open reading frame filtering methods" which is essential step needed for protein-protein interactions. Following the internship, she joined Malek's lab as a Research Specialist and was involved in the research for all versus all interaction mapping "AVA-seq" a novel method for identifying protein-protein interaction.

NOHA YOUSRI

Assistant Professor of Research in Genetic Medicine
Weill Cornell Medical College, Qatar

Noha A. Yousri has joined the Bioinformatics Core in Weill Cornell Medicine- Qatar (WCM-Q) in 2012. She is currently a Research Associate in the department of Genetic Medicine department in WCM-Q, and has long been doing association studies with different phenotypes including T2D and Arthritis among others. She has lead the computational analysis for several studies on genomics and metabolomics data including GWAS studies. She comes from a Computer Engineering background, with a specialization in machine learning techniques. She is also affiliated with Alexandria University as an Associate Professor of Computer and Systems Engineering. She has authored and co-authored more than 30 peer reviewed publications in the fields of computer engineering, bioinformatics and biological/medical sciences.

SAHAR DAAS

Laboratory Supervisor
Sidra Medicine

She is currently the laboratory supervisor of Zebrafish Core Facility at Sidra Medicine where she has established zebrafish model to be used for functional genomics to validate and characterize the phenotypes of genetic variants identified in the Qatar Genome Program related to inherited diseases that are predominant in Qatar. She possesses significant experience in the laboratory operation and her commitment is to develop an advance level of laboratory services. Currently she is finalizing her PhD at the Hamad Bin Khalifa University, her research project is The Role of Cardiac Myosin Binding Protein C3 Variants in Hypertrophic Cardiomyopathy: Correlation of Genotype-Phenotype in the Zebrafish Model. Prior relocating to Qatar, she worked at the Berman laboratory in Canada. Her research focused on the characterization of the notch signaling pathway role in mast cell development aiming to identify novel therapeutic strategies for targeting mast cells disease. The results of this work have been published in scientific articles and book chapter. Sahar has received awards for excellence in research including the professional excellence Thana award- Qatar foundation, the innovation excellence award – cancer care nova scotia, Canada and the IWK hospital stars team recognition award, Canada and more.

SARA AL-KHAWAGA

Dermatology and Venereology Resident
Hamad Medical Corporation

Dr. Sara Al Khawaga obtained her medical degree with several honors from Weill Cornell Medicine-Qatar in 2014 where she continually demonstrated excellent qualities and outstanding performance. She later joined Hamad Medical Corporation for her transitional year of residency in 2015. Sarah demonstrated great interest in clinical and translational research, she later joined Sidra Medicine-Qatar to complete a postdoctoral fellowship. Concurrently, Dr. Sarah has been enrolled in Hamad Bin Khalifa University (HBKU) to pursue her PhD in Biological and Biomedical science. The focus of her PhD thesis is on using human pluripotent stem cells including embryonic stem cells (hESC) and induced pluripotent stem cells (iPSCs) to study the role of a specific pancreatic transcription factor (TF) during pancreatic beta cell differentiation and studying genomics of Neonatal Diabetes in The State of Qatar.

SARMAD ALI GHULAN SHABBIR

Laboratory Senior Technologist
Hamad Medical Corporation

He received Masters in Biochemistry in 2009 from the University of Karachi, Pakistan. He joined Aga Khan University Hospital in 2010, where I got training in the field of Cytogenetics and Molecular Genetics.

At Aga Khan University Hospital he worked for three years in the areas of Cell culture, Fluorescent in situ Hybridization (FISH) and Chromosomal analysis on wide range of samples such as bone marrow, peripheral blood, amniotic fluid, CVS, Skin biopsy and Product of conception.

He joined Hamad Medical Corporation in 2013 and currently working as Senior Technologist in the areas of Microarray, Cell isolation techniques and NGS based Non-invasive prenatal testing (NIPT).

ZOHREH CALDERONE

Staff Scientist
Sidra Medicine

Dr. Tatari-Calderone earned her Ph.D at the University of Paris Diderot/Saint Louis Hospital in Paris, France in the laboratory of Nobel Laureate Dr. Jean Dausset, under the direction of Drs. Dominique Charron and Elian Gluckman. She developed a new technique for HLA-Cw genotyping to evaluate its role in the severity of graft versus host disease (GvHD) after unrelated bone marrow transplantation (UBMT).

She then joined the laboratory of Dr. Ron Gress in the Experimental Transplantation and Immunology Branch (ETIB) at the National Institutes of Health (NIH) as a postdoctoral fellow, where she worked on the role of IL-15 in hematopoietic reconstruction after BMT or chemotherapy.

About Sidra Medicine

Sidra Medicine provides specialized healthcare to women, children, and young people from Qatar and around the world. It is a private hospital for public benefit.

Sidra Medicine represents the vision of Her Highness Sheikha Moza bint Nasser who serves as its Chairperson. This high-tech facility not only provides world-class patient care, but also helps build Qatar's scientific expertise and resources.

Established by Qatar Foundation for Education, Science and Community Development, Sidra Medicine embraces best practice medical education, innovative biomedical and clinical research and discovery, and exceptional patient and family focused care. This unique combination makes Sidra Medicine one of the handful of healthcare organizations in the world that embeds the concept of personalized medicine into its very ethos of treatment and care.

Sidra Medicine provides comprehensive specialist healthcare services for children and young people, as well as maternity and gynecology care for women. Some of its unique children's specialist services include Cardiology, Neurology, Urology and Plastics & Craniofacial Reconstruction. It is also treats and cares for pregnant women with fetal complications.

The ultramodern healthcare organization is a testament to Qatar's pioneering spirit and sustained commitment to its human and social development. To access Sidra Medicine services, and learn more about our contribution to global healthcare, education and research, please visit www.sidra.org.

OUR COMMITMENT TO MEDICAL SCIENCE ADVANCEMENT

Sidra Medicine is also part of a dynamic research and education environment in Qatar and through strong partnerships with leading institutions around the world, we are creating an intellectual ecosystem to help advance scientific discovery through investment in medical research.

Based in Education City and as an entity of the Qatar Foundation for Education, Science and Community Development (QF), Sidra Medicine is part of a dynamic research and education environment. Education City includes leading international institutions such as Weill Cornell Medicine-Qatar, Georgetown University, Carnegie Mellon University, and HEC Paris as well as recently established national research centers like Qatar Biomedical Research Institute and Qatar Environment and Energy Research Institute.

We work closely with academic partners like Weill-Cornell Medicine in Qatar (WCM-Q) and Qatar University (QU) as well as other health and research institutions like Hamad Medical Corporation (HMC), Primary Health Care Corporation (PHCC) and Qatar Biobank in regard to all three missions – raising the standard of healthcare throughout the country and providing valuable opportunities for research and learning.

THE SIDRA TREE STORY

Qatar Foundation's logo is the Sidra tree (Ziziphus spina-christi) which is native to Qatar and flourishes in the harsh and arid climate. It grows in the wild and can be found throughout the country, especially in northern and central Qatar.

QF consists of around 50 centers and the branches of the Sidra tree represent the diversity of QF today. The leaves, flowers and fruits equate to the individual lives that the tree nourishes, with the fruits going on to produce seeds that guarantee sustainability and a healthy future.

The Sidra tree's deep roots are a strong anchor, connecting contemporary learning and growth with the country's culture and heritage.

Poets, scholars and travelers would traditionally gather in the shade of the Sidra's spreading branches to meet and talk. This aspect of the Sidra tree's role is reflected in QF's commitment to education and community development. As well as being a naturally comfortable and convenient place at which to gather and exchange knowledge and opinions, it is also a very healthy location. The tree's fruit, flowers and leaves provide the ingredients for many traditional medicines, which reflects QF's science and research objectives.

The Sidra tree is perhaps the most prominent tree in Qatar and it means many different things to many different people. It certainly occupies a special position in the hearts of the Qatari people, which is why it is the perfect symbol for the vision and mission of QF today.

About Qatar Foundation

VISION

Through education and research, Qatar Foundation leads human, social, and economic development of Qatar; making Qatar a nation that can be a vanguard for productive change in the region and a role model for the broader international community.

MISSION

Qatar Foundation for Education, Science and Community Development is a private, non-profit organization that serves the people of Qatar by supporting and operating programs in three core mission areas: education, science and research, and community development. The Foundation strives to nurture the future leaders of Qatar. By example and by sharing its experience, the Foundation also contributes to human development nationally, regionally, and internationally. In all of its activities, the Foundation promotes a culture of excellence in Qatar and furthers its role in supporting an innovative and open society that aspires to develop sustainable human capacity, social, and economic prosperity for a knowledge-based economy.

About Qatar National Vision 2030

During the reign of His Highness Sheikh Hamad bin Khalifa Al Thani, the Father Emir, May God Protect Him, Qatar National Vision 2030 was launched to serve as a clear roadmap for Qatar's future. It aims to propel Qatar forward by balancing the accomplishments that achieve economic growth with the human and natural resources. This vision constitutes a beacon that guides economic, social, human and environmental development of the country in the coming decades, so that it is inclusive and helpful for the citizens and residents of Qatar in various aspects of their lives.

The Qatar National Vision 2030 defines broad future trends and reflects the aspirations, objectives and culture of the Qatari people. By shedding light on the future, the Vision illuminates the fundamental choices that are available to Qatari society. Simultaneously, it inspires Qatari people to develop a set of common goals related to their future.

Qatar's National Vision defines the long-term outcomes for the country as a whole rather than the processes for reaching these outcomes. It provides a framework within which national strategies and implementation plans can be developed.

The National Vision aims at transforming Qatar into an advanced country by 2030, capable of sustaining its own development and providing for a high standard of living for all of its people for generations to come.

Visitors Guide

HISTORY

Archaeological evidence show human habitation of Qatar dating back to the Stone Age, and that there was a healthy seagoing community in the peninsula as early as 5000 BC. Hunting, gathering and fishing supported these early communities.

In medieval times, Qatar was an important trading post in the Gulf-Indian Ocean commerce. It was dominated by the Ottomans in the 19th century and then became a British protectorate in 1916. The arrival of oil prospectors and the establishment in 1935 of Petroleum Development Qatar signaled the beginning of a new era. Since gaining independence in 1971, the sovereign state has transformed itself into an economic powerhouse.

The ruling Al Thani family, a branch of an ancient Arab tribe, settled in Qatar during the early 18th century. The current Emir is His Highness, Sheikh Tamin Bin Hamad Al Thani.

Despite ongoing 21st century advancements, Qatar places great value on its most important possession, its rich cultural heritage. Customs and traditions are still widely observed and historical sites are well-preserved.

BLEND OF OLD AND NEW

The stunning urban landscape of Doha can make one forget that the beginnings of Qatar date back 7,000 years. Qatar always fascinates with its contrasting, open, 21st century society that is rooted in culture and history. It has soaring skyscrapers and beautiful mosques, local souqs and upscale malls, falcons homing back to their masters, jets that cut through the sky, historic forts and futuristic hotels, camel races and international sporting events.

KEY INDUSTRIES

Oil and gas production are the drivers of Qatar's economy. Oil production capacity is 850,000 barrels per day. Qatar is the largest global exporter of liquefied natural gas (LNG) producing some 38 million tons annually. Efforts to diversify the economy are high on the agenda. The result is a broad range of growth industries including banking, telecommunications and IT, fertilizer production, aluminum smelting, construction, tourism, and real estate.

CULTURE

Qataris are passionately committed to upholding their heritage and cultural values while forging one of the most advanced societies in the world. Its ancient history can be glimpsed in carefully preserved heritage sites including forts and prehistoric settlements where rare petroglyphs have been discovered.
In the metamorphosing capital city of Doha itself, the passion for capturing and reviving Qatar's cultural heritage is felt everywhere – magnificent museums housing priceless artefacts, restored Bedouin souqs, merchant houses, and royal residences, prestigious races and shows purely for Arabian horses, and traditional wooden dhows plying the bay.

CLIMATE

The climate is characterized by a mild winter and a hot summer. Rainfall in the winter is slight, averaging some 80 millimeters a year. Temperatures range from 7 degrees centigrade in January to around 45 degrees at the height of summer (July and August). The weather is generally pleasant from October until May.

Safety & Security Doha is recognized as one of the safest cities, and Qatar as one of the safest countries in the world. Business travelers arriving alone or with their families for the Symposium can take confidence in Qatar's 2014 ranking by the Global Peace Index as the 22nd most peaceful of 162 nations of the world. It is the only country in the Middle East to be ranked among the top 20 most peaceful nations. Qatar's crime rate in all categories is a fraction of the world average – one quarter of the average for robberies per 100,000 people and 12.5% of the average for homicides. The government has ensured that residents and visitors of all backgrounds and nationalities feel at home and safe in Qatar. Thus, Americans, Brazilians, British, Dutch, Filipinos, Germans, Indians, Japanese, Lebanese, and South Africans, to name but a few, live and work together in harmony. United Nations agencies frequently hold international meetings and conferences in Doha, due in great measure to its peaceful and secure setting.

CULTURAL EXPERIENCES

Qataris are passionately committed to upholding their heritage and cultural values while forging one of the most advanced societies in the world.

Accessibility and transportation

TRAVEL INFORMATION

Qatar's unique location close to the Indian, Southeast Asian and Far East economic powerhouses, as well as the consumer markets of the west, has set the stage for this new global crossroads. Qatar is only six hours away from the capital cities of Europe and Asia and 13 hours from major destinations in North America. Doha International Airport is served by 35 international airlines which all operate regular scheduled flights from Europe, the U.S. and the Asia-Pacific region among others. The airport is just five kilometers from the city center. Qatar Airways is leading the race in redeveloping the region's future aviation hub, the New Doha International Airport. Situated approximately four kilometers east of the existing airport, the new facility will be the world's first airport to accommodate unrestricted operations by all commercial aircraft, including the A380 – the largest passenger aircraft ever built.

Qatar Airways operates a modern fleet of 119 Airbus and Boeing aircraft and with over 250 more on order. Qatar Airways is one of the world's fastest growing carriers. It is one of only six airlines to be awarded five – star status by Skytrax and is consistently ranked as the Middle East's top airline rated Best Middle East Airline for six consecutive years and named Airline of the Year 2011 and 2012 by Aviation Business. Qatar Airways currently flies to more than 120 diverse business and leisure destinations across Europe, the Middle East, Africa, South Asia, the Far East and North America. The airline's code-share partners include All Nippon Airways, Asiana Airlines, Gol Linhas Aéreas Inteligentes, Malaysia Airlines, Middle East Airlines, Philippine Airlines, US Airways, Azerbaijan Airlines, SNCF and Oman Air.

Other international airlines delegates also benefit from the air services of nearby hubs Abu Dhabi and Dubai in the United Arab Emirates (UAE). Both cities are less than one hour away from Doha and offer additional connections to international destinations, further enhancing Qatar's global access. European and African carriers operate direct services to Doha and regional hubs. U.S. national airline Delta flies from Atlanta to Dubai and United flies from Chicago to Dubai. US Airways is Qatar Airways' partner in USA and connections can be made via New York, Washington, Houston and Montreal from any city in North America. Qatar Airways currently flies to more than 120 destinations directly covering all regions.

TRANSPORT

The national transport company Mowasalat operates bus, taxi and limousine services. Taxi service is 24 hours; taxis can be hailed from the side of the road. The fare from the airport to any destination in Doha by cab is approximately QAR40 (US$11). Limousines can be booked for specific journeys or by the hour.

Visitor information

ALCOHOL

Qatar has a relatively liberal attitude to the consumption of alcohol for non-Muslims. Liquor is available in many hotels. However it is strictly forbidden to bring alcohol into the country including Duty Free. During the Holy month of Ramadan alcohol is not served.

BANKING

Automatic Teller Machines (ATMs) are readily accessible throughout the city. Banking hours are generally 7.30am – 1pm Sunday to Thursday with some branches open in the afternoons.

BUSINESS HOURS

The working week in Qatar runs from Sunday to Thursday with Friday (the holy day for Muslims) and Saturday being days off.

COMMUNICATION

Local telephone calls are free. The majority of hotels offer broadband connectivity various cafes and parks around Qatar offer free wireless and wireless "hot spots" are becoming more frequently accessible.

COMMON COURTESIES IN QATAR

Women are highly regarded in business at all levels. Western men and women freely wear western style clothing. However it is advisable that clothing for both men and women covers the knees and sleeveless tops showing the shoulders and upper arms should be avoided. Tight or revealing clothing is unseemly for both men and women. Arab women are very modest so it is recommended that men approach other males for directions or advice.

EMERGENCIES

Emergency Services in Qatar can contacted by calling 999

PHOTOGRAPHY

It is requested that permission is sought before photographing anyone including local Qataris.

EMBASSIES

Qatar has 78 diplomatic embassies and six general consulates abroad with over 50 foreign Diplomatic Representatives in Qatar.

GOVERNMENT
The Emir is the ruler of Qatar. Rule is hereditary within the family of Al Thani whereby power is transferred from father and son. The Emir is the head of the constitutional authorities holding both legislative and executive powers. The Emir appoints the Prime Ministers and Ministers. The Council of Ministers (Cabinet) the supreme executive authority assists in implementing the general policies of the State.

NEWSPAPERS
There are three English daily newspapers in Qatar: Gulf Times The Peninsula and the Qatar Tribune.

POPULATION
1 670 389 (Qatar Statistics Authority – May 2011)

RESOURCES
Primarily petroleum and gas although finance – insurance – real estate – business services – manufacturing industries – medical care and research are all growing sectors.

SAFETY
Qatar is ranked as one of the safest places in the world by the Global Peace Index above France UK and USA. Strolling the corniche or walking around the city center walking to and from your hotels is a very safe environment.

SMOKING
Qatar has tobacco-control laws banning smoking in public places such as restaurants shopping centers and sports venues. Many consider smoking “shisha” the perfect complement to local food and drink. The pipe is filled with water and tobacco which is available in different flavors such as apple strawberry and even chocolate.

TRAVELERS WITH DISABILITIES
Many of the hotels have ramps allowing for wheelchair access into the building and elevators once inside. The larger more modern hotels have rooms for disable guests. Most of the malls have elevators and escalators and enlarged washroom facilities for disabled guests. Al Maha Doha’s Meet & Greet Service can offer assistance to physically challenged visitors upon arrival.

VISA
A visa is required and nationals from more than 30 countries will be issued with a visa upon arrival. Visas can be pre-arranged electronically through the official Qatar government website. Passport holders of countries unable to receive a visa on arrival can arrange with their hotel.

WATER
Water in Qatar is safe to drink. Many people prefer to drink bottled water.

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