مؤسسة قطر
Qatar Foundation

سدرة للطب
Sidra Medicine

SIDRA MEDICINE
RESEARCH

Annual Report 2020

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Welcome Note
from the Chief Research Officer

In many ways, 2020 was a most challenging year: the sudden onset of a global pandemic, the burden on healthcare facilities, and the anxieties we all had to endure made adapting to pandemic life very stressful. Yet ironically, despite the increases in physical distance, never in our lives did humanity come so closely together to face a common enemy. And little did we know that a global pandemic, challenging as it may be, would be a blessing in disguise for science... with the world's attention fixated on biology and public health, terms once considered esoteric jargon – R0, Ct value, mRNA, PCR, antigens, etc. – entered the common vernacular! ... and scientific inquiry captured the public's imagination.

Playing to its strength as a national academic medical center, the Sidra Research Branch made significant achievements during the pandemic. On one end, our Core Genome Laboratory developed methods to test for viral infection in nasopharyngeal and saliva samples which cleverly bypassed global shortages for testing reagents. On another, our teams of geneticists and immunologists joined local and global consortia investigating penetrance and variability of Covid19 in adults and children, leading to discoveries that impacted clinical care worldwide. As vaccines begin to roll out, our Deep Phenotyping Core developed an innovative serology assay, which detects antigens to a whole range of human coronaviruses, and could serve as an immunity passport as the global economy re-opens. In addition to Covid19 projects, the Research Branch had a remarkably successful year. Over 1,100 patients and families enrolled in research studies in 2020. Sidra clinicians and scientists published nearly 300 papers, with almost 75% of all Research Branch publications in the top 15% of international journals. Importantly, Sidra Medicine was awarded 18 national grants, demonstrating the growing recognition of the importance of academic medical centers for translational discovery.

In terms of contribution to Qatar's knowledge economy, Sidra continued to develop trainees in the fields of medicine and biomedical research. Almost 60% of Research Investigators have academic appointments at local and international institutions, and a total of 47 trainees and graduate students were part of the Sidra Research family in 2020. Sidra Research also maintained a 20% Qatarization rate, supporting growth and development of the brightest talent to lead this growing field in the future. Finally, Sidra Research hosted its annual flagship 'Precision Medicine and Functional Genomics' symposium virtually this year, demonstrating our commitment to education and sustainability, and to building a research enterprise in Qatar with strong links to the global scientific community.

In summary, the Sidra Research 2020 Annual Report covers what ultimately became one of the most productive and innovative years at the Research Branch. We owe a most sincere gratitude to our hard-working research staff who braved the pandemic and worked around the clock to ensure discovery and innovation remained strong, to our diverse clinical collaborators who were fundamental in devising studies and translating outcomes to patients, and to all the families and patients who entrust Sidra Medicine to deliver the highest-quality, research-driven care. We hope you enjoy this report, and look forward to a 2021 filled with terrific achievements and outstanding discoveries.

Dr. Khalid A. Fakhro
Chief Research Officer

Research **Mission** and **Strategy**

Dr. Khalid A. Fakhro
Chief Research Officer

Dr. Ziyad M. Hijazi
A/Chief Medical Officer

In line with its vision to be a world renowned academic medical center, Sidra Medicine is built upon three mission-critical pillars: education, research, and clinical care. The Research Branch at Sidra is tasked with two strategic goals: establishing a strong, clinically oriented biomedical research program, and developing a national resource of genomic information that improves health in Qatar and the region. Achieving these goals requires significant collaboration both internally – among research, corporate and clinical stakeholders – and externally, via strategic partnerships with other Qatar-based and international institutions, that combine to establish world-class, research-driven care for our patients.

In line with these goals, the Research Branch is tasked to deliver a hospital-wide Precision Medicine Program for Sidra, built upon the philosophy that research technologies and innovation should play a prominent role in every patient's journey at Sidra. Strategically, this Program is designed to integrate research into clinical workflows across three pillars:

1. Research for every patient – developing protocols that allow every patient access to cutting-edge research studies via informed consent
2. Advanced Diagnostics – leveraging next-generation research technologies (e.g., genome sequencing) to dissect diseases and identify their causes at an ultra-high resolution, beyond the limits currently available as standard-of-care
3. Personalized Therapy – Establishing infrastructure and systems to deliver advanced therapies (e.g., stem cells and gene therapy) and support a culture of clinical trials at Sidra

These strategic pillars require close collaboration, trust and interactions between patients, trainees, physicians, and researchers to advance care at Sidra. Such a multidisciplinary approach reinforces the culture of OneSidra, where education, research and clinical care merge seamlessly and become part of every patient's journey, ensuring that discoveries made from patients go towards improving their very own care, delivering personalized medicine to all.

In summary, we recognize that the implementation of these pillars into patient care is not easy, however, we view this as a common goal that will become a key differentiator, setting Sidra Medicine apart from its peers in the local, regional and international context, and positioning Sidra Medicine as a unique Academic Medical Center serving patients from Qatar and abroad.

Dr. Khalid A. Fakhro
Chief Research Officer

Dr. Ziyad M. Hijazi
A/Chief Medical Officer

Leadership **Structure**

Chief Research Officer

Khalid A. Fakhro
Director, Human Genetics Department

Deputy Chief Research Officer

Rashid Al Ali
Division Chief, Bioinformatics and Digital Health

Scientific Department Directors

Damien Chaussabel
Immunology

Davide Bedognetti
Cancer

Souhaila Al Khodor
Maternal and Child Health

Core Directors

Jean-Charles Grivel
Deep Phenotyping Core

Stephan Lorenz
Integrated Genomics Services

Nasser Elkum
Biostatistics and Clinical Epidemiology

Chiara Cugno
Advanced Cell Therapy Core

Max Renault
Research Operations and Services

Internal **Research Council**

The Internal Research Council develops Sidra Medicine's translational research roadmap, ensuring it remains internationally competitive. One of the central goals of the IRC is strengthening Clinical-Research integration and transforming Sidra Medicine to be among the best institutions in the world offering research-driven care.

The IRC oversees Sidra Medicine's progress towards delivering its long-term strategy for Research. It achieves this by evaluating projects for scientific merit, academic rigor, alignment to national and clinical priorities, and overall impact to patient care. In this way, the IRC ensures Sidra Medicine's Research agenda closely reflects national priorities while establishing a long-term culture of "oneness" across Sidra Medicine's three strategic pillars: research, education and patient care.

The IRC is chaired by the Chief Research Officer who along with six senior research scientists and clinician scientists set research priorities for Sidra Medicine. All members were jointly nominated by the Chief Research Officer and the Chief Medical Officer based on a seniority and track record of high-impact research experience and a competitive research funding track record. The members have demonstrated a dedication to Sidra Medicine's values of teamwork, efficiency, care, and innovation necessary to deliver a world-class research program at Sidra Medicine.

Chairperson
Khalid A. Fakhro
Chief Research Officer

Vice Chairperson
Ibrahim Janahi
Division Chief, Pediatric Pulmonology

Member
Damien Chaussabel
Director, Immunology Department

Member
Davide Bedognetti
Director, Cancer Department

Member
Khalid Hussain
Division Chief, Endocrinology

Member
Souhaila Al Khodor
Director, Maternal and Child Health Department

Member
Colin Powell
Senior Attending Physician, Emergency Department

Scientific **Departments**

Human Genetics

Dr. Khalid A. Fakhro
Director, Human Genetics

The Human Genetics Department is led by Dr Khalid A. Fakhro. Dr. Fakhro graduated from the University of Chicago (USA) majoring in Biological Sciences with specialization in Cellular Biology and Molecular Genetics, and later completed his PhD in Human Genetics at Yale University (USA), where he was part of a highly selective HHMI Translational Medicine Scholars program, designed to train basic scientists to take discoveries from the bench to bedside. After two post-doctoral training fellowships abroad, he relocated to Qatar as part of the Department of Genetic Medicine at Weill Cornell Medical College in Qatar. He subsequently joined Sidra as Principal Investigator, and became the Director of the Human Genetics department, where he built a robust genomic medicine research pipeline for the hospital. In 2018, he became the inaugural Director of Human Genetics at Sidra, heading an ambitious program where research in genomics and personalized medicine are embedded in the heart of Sidra Medicine's academic medical enterprise.

The Human Genetics Department focuses on two pillars: advanced diagnostics of patients with genetic disease, and the characterization genomic variation within the Qatari population. The diagnostic track of the department aims to address key questions related to disorders affecting Sidra patients, including understanding what genetic mutations may be causing disease (or susceptibility to disease), and how can this knowledge benefit patient care. Importantly, scientists in the department go beyond identification of genetic variants to investigating the molecular mechanisms of disease. This is done using a variety of tools, including: cellular models, zebrafish and stem cells, etc. These models help scientists better understand what drives disease in patients, opening a window to more targeted treatment.

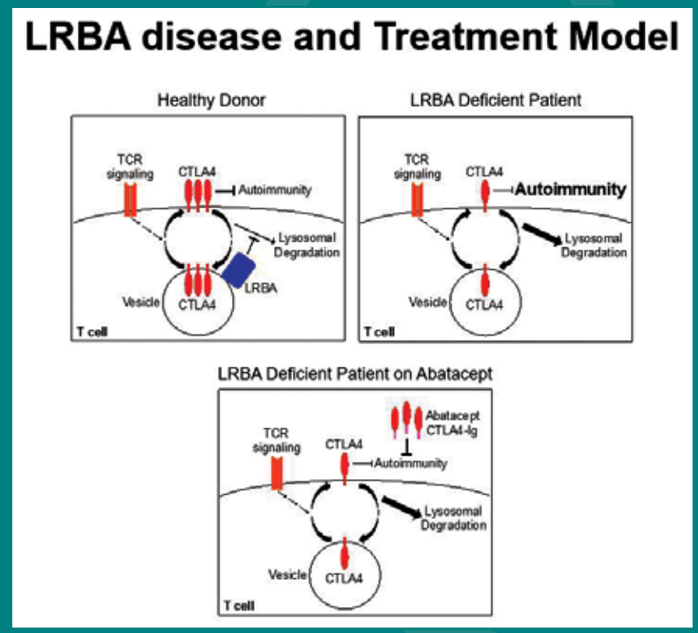
In addition to developing advanced diagnostics and therapy at Sidra, the department of Human Genetics plays an important role in characterizing genomic variation among Qataris through population genetics studies. This includes studies of population structure, that have identified several key ancestries that make up the fabric of Qatari society, and uncovered different disease susceptibilities that may be higher in one group versus another due to genetic differences. Further, the Human Genetics Department seeks to map the structure of the Qatari genome, which has been shown to harbor a greater diversity than originally anticipated. Finally, population level analysis also uncovers signatures of the pressures that ancestors of modern day Qataris had to face, including identifying variations in the expression of certain genes that make the population more or less susceptible to diseases today.

The Human Genetics Department is designed to offer a strong research department in understanding the genetic underpinnings of disease in the context of population-scale variation, to producing high impact research discoveries that change patient lives. Importantly, this is achieved with an eye on building capacity through developing the next generation of young talented scientists, who ensure long-term sustainability in research on Genetics and Precision Medicine in Qatar.

Human Genetics Highlight

An Effective Targeted Therapy for LRBA-deficient Patients

Investigators from the Human Genetics department work closely with clinicians at Sidra Medicine. The Allergy and Immunology Department at Sidra, which specializes in inherited immune deficiency disorders, recently diagnosed early onset autoimmune disease patients with mutations in a gene called LRBA. When LRBA mutations were first discovered in 2012 to cause an inherited autoimmune disease, the function of LRBA and how it caused disease was mostly unknown. Research investigations from Dr. Bernice Lo and her colleagues helped determine that LRBA regulates the levels of another protein called CTLA4. CTLA4 is a crucial immune regulatory molecule that helps prevent autoimmune disease development.



Disease mutations in LRBA caused the loss of LRBA protein and were found to result in the loss of CTLA4 protein as well, thus leading to autoimmune disease. A recombinant CTLA4 drug called abatacept that is used to suppress autoimmunity in rheumatoid arthritis was found to show efficacy in treating LRBA deficient patients. Since LRBA deficiency leads to loss of CTLA4, abatacept acts as a targeted replacement therapy. Generally, treatment of children with LRBA deficiency includes trial of different immune suppressive therapies and Hematopoietic Stem Cell Transplant (HSCT) as potentially curative option. The outcome is poor with significant morbidity and mortality.

Due to the research conducted by the Human Genetics department investigators, Dr. Amel Hassan treated her LRBA-deficient patients with abatacept and found that the personalized therapy has significantly improved the patient clinical condition reflected by significantly less infections in both frequency and severity. Inflammation was also much more controlled with less gut symptoms and patient's weight gain is remarkably improved. Significant reduction in admission to hospital was also noted in terms of both frequency and length of stay.

Scientific **Departments**

Immunology

The Immunology Department is led by Dr. Damien Chaussabel. Dr. Chaussabel is a trained immunologist, and has acquired expertise in the genomics and bioinformatics fields. Prior to joining Sidra, Dr. Chaussabel developed a genomics and bioinformatics program at the Baylor Institute for Immunology Research in Dallas TX. He served as head of the Systems Immunology Division at the Benaroya research institute in Seattle WA where he led studies investigating "genomic reprograming" that occurs in the blood of patients with infectious and autoimmune diseases as well as in response to vaccination.

Dr. Damien Chaussabel
Director, Immunology Department

Immunity plays an important role in health and disease. In patient studies the immune system has traditionally been investigated in the context of the immunological processes that are responsible for, allergic, auto-immune and infectious diseases. But more recently its role has become more widely recognized in the pathogenesis of diseases, such as cancer or metabolic disfunction.

Specifically, the objectives of the Immunology department at Sidra Medicine are: 1) To identify immune pathways associated with disease pathogenesis and health maintenance; 2) to foster the development advanced immune profiling / diagnostic modalities and 3) to help guide treatment with an increasingly wider range of immune modulating drugs and therapies.

The Immunology department is organized as a multi-disciplinary group that employs cutting-edge cellular and molecular profiling approaches in the context of patient-based studies. The department is comprised of investigators whose expertise ranges from immunogenetics, tumor immunology, molecular and cellular immunology to systems immunology.

This expertise is currently being applied towards addressing the clinical needs of patients with rare inborn errors of immunity, as well as patients with more common diseases, such as asthma, allergy and diabetes. It also strives to foster research activities in combination with clinical units, including Sidra's neonatal intensive care, pathology, allergy and clinical immunology, pediatric pulmonology, pediatric infectious diseases, as well as the obstetrics and gynecology departments.

Overall, current activities within the department have led to the development of advanced immune profiling platforms. Relying on cutting-edge profiling technologies, these platforms allow us to assess the immune status of individuals with unprecedented levels of details. Research projects are also implemented that aim to associate these high-resolution molecular and cellular phenotypes with health states and outcomes, to ultimately permit the implementation of "precision immunology" approaches at the bedside.

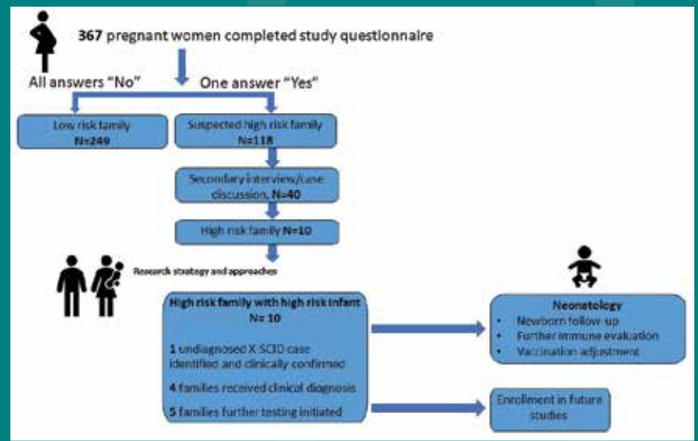
Immunology Highlight

Non-invasive Screening of Patients with High Risk to Life-threatening Infections

Primary Immunodeficiency Diseases (PIDs) are classified as inherited errors of immunity leading to a variety of clinical symptoms, including infections, inflammatory and/or autoimmune diseases. They are more prevalent in populations with high rates of consanguinity; however, many patients are diagnosed late, or a genetic diagnosis remains elusive.

Dr. Nico Marr and his team in the Immunology department hypothesized that simple questionnaire-based screening may identify undiagnosed PID cases among newborns, thereby allowing personalized clinical management and reducing the risk of life-threating complications due to the administration of live vaccines and severe infections early in life. Investigators surveyed pregnant women, who attended antenatal care at Sidra, about age, nationality, pregnancy history, consanguinity and occurrence of severe infections or side effects to vaccinations in the extended family.

Potential high-risk families received an additional consultation by Immunology/ Allergy and Neonatology Clinics in Sidra, to consider further clinical assessment, including immunological work up of the newborn and/or delayed live attenuated vaccination. A total of 367 pregnant women (median age: 31, range: 19-45) were consented and interviewed, including 141 Qatari nationals.



The frequency of consanguinity was self-reported at 22% of cases and previous pregnancy history was split almost equally in nullipara, primipara and multipara. Percentages of pregnant women with an older child or a family member who suffered from any of the following were reported: severe and/or repeated infections (9%), severe side-effects to vaccination (7%), severe allergies (20%) and known or suspected PID diagnosis (5%).

About 40 women were followed up by a secondary interview and/ or immunology case discussion. Of them, ten families were subjected to whole genome sequencing (WGS). Four families received a clinical diagnosis during the study and were not followed up further. For at least five families, further clinical testing was initiated as a result of our study.

Thus far, researchers have identified an undiagnosed X-SCID case, which was clinically confirmed. The investigations and data analysis for the other high-risk children is currently ongoing, with a suspected underlying PID diagnosis in at least two more families that were sequenced. Based on the findings, investigators strongly recommend incorporating a similar screening as part of the routine antenatal care in Qatar to allow for a more personalized medicine approach in the future.

Scientific **Departments**

Cancer

Dr. Davide Bedognetti
Director, Cancer Department

Dr. Davide Bedognetti, is the Director of Cancer department at the Sidra Medicine. He received his MD and PhD in Clinical and Experimental Oncology and Hematology from the University of Genoa, Italy. After obtaining the Board Certification in Medical Oncology by the University of Genova and Italian National Cancer Institute (IST), he joined the Infectious Disease and Immunogenetics Section (IDIS) of the US National Institutes of Health (NIH) where he completed his post-doctoral fellowship. He has also served as Director of the Federation of Clinical Immunology Societies (FOCIS) Center of Excellence at NIH Clinical Center. Dr. Bedognetti is member of the Society for Immunotherapy of Cancer (SITC) Cancer Immune Responsiveness Taskforce, and the FOCIS Centers of Excellence Steering Committee.

Cancer is the leading cause of death by disease in children and adolescents and the second leading cause of death in adults globally. The implementation of personalized therapeutic strategies is hindered by the incomplete knowledge of the molecular mechanisms driving heterogenous types of cancer in different individuals. Cancer immunotherapy is revolutionizing the way patients are treated but only a minority of patients respond to such as a treatment.

The Cancer Research department aims to characterize each patient and his/her tumor at the deepest level of complexity to define hierarchically relevant alterations that can be targeted by specific approaches.

Conceptually, the department can be divided in two parallel, yet complimentary, tracks: Discovery and Clinical implementation:

The Discovery track: The department aims to address the key questions related to precision medicine in cancer patients, with an emphasis on cancer immunotherapy. In essence, it aims to understand the molecular mechanisms associated with neoplastic development, treatment resistance, and treatment toxicity, and to perform pre-clinical, proof-of-principle studies on novel therapeutic approaches.

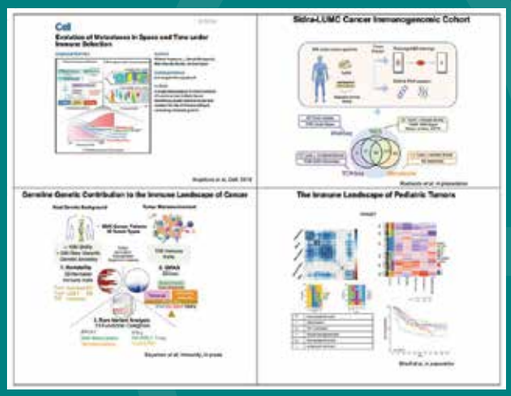
These findings might be used to refine stratification systems, therefore allowing personalized treatments, and to implement novel therapeutic approaches in both adult and pediatric settings. Samples used for discovery purposes come from Sidra and from collaborative local and foreign institutions or consortia.

The Clinical Implementation track: It is expected to put into practice the concept of precision medicine in pediatric cancer patients treated at Sidra, also implementing in the clinical setting the findings generated by the Discovery component. Activities of the department will be complemented with the ones from orthogonal departments such as the Human Genetics, Immunology and Maternal and Child Health departments.

Cancer Highlight

Lack of Immune Response Predicted the Risk of Tumor Relapse

Can we predict tumor relapse or progression in metastatic cancer patients by assessing the level of the immune response?



Using state-of-the-art technology available at Sidra Medicine paired with integrative analytic pipelines, Cancer department investigators directed by Dr. Davide Bedognetti provided the most comprehensive immunogenomic analysis of tumor metastases available so far. They demonstrated that tumor clones seeding other metastatic sites are “invisible” to the immune system and characterized this phenomenon at the genetic level. For each metastasis, scientists computed an immunoediting score that captures the level of immunologic pressure. Clones that did not relapse were typified by a high immunoediting score, while the ones that persist or relapse displayed a low level of genetic immunoediting.

The combined immunoediting score of different metastases was used to predict the risk of relapse in two patients. In patient A, the model predicted less than 35% of risk of relapse before January 2018. In patient B, two resected metastases had an estimated risk of relapse of 95% and 99%. This analysis was performed blinded, without access to the clinical information. Indeed, patient A was still alive at the date of publication (October 2018) while patient B developed several metastases conducive to death. These findings have a profound effect on personalized medicine research worldwide. If alterations associated with a high risk of death are identified, these could be targeted to avoid that tumor generate metastases.

Through a recently-awarded QNRF-NPRP grant, Sidra Medicine’s researchers in collaboration with INSERM and Hamad Medical Corporation (HMC) scientists and clinicians, plan to expand this approach to a larger number of metastatic patients. The immuno-editing score might be used to predict the chance to relapse and to define personalized treatment in high risk patients. In addition, the investigators recently defined a specific immune signature able to predict cancer recurrence with superior precision as compared to conventional clinicopathologic and molecular parameters, and, in collaboration with The Cancer Genome Atlas they proposed a cancer classification based on immunologic features derived from genomic analysis. Similar approaches can be applied to pediatric tumors to understand the basis of disease progression and identify novel targets. Together with Dr. Catherine Cole, Division Chief of Pediatric Hematology, and Dr. Ayman Saleh, Oncology and Hemopoietic Stem Cell Transplantation, Cancer department investigators are launching a Precision Medicine Cancer program focusing on Sidra’s patients.

This is a common initiative involving clinicians and scientists. The aim will be able to provide a deep molecular characterization of each single patient in order to inform therapeutic decision and to gain knowledge about mechanisms involved in disease development.

Scientific **Departments**

Maternal and Child Health

Dr. Souhaila Al Khodor
Director, Maternal and Child Health Department

Dr. Souhaila Al Khodor received her bachelor's degree in medical Laboratory technology from the Faculty of Public Health at the Lebanese University in 2001. Soon after, she started her Master's 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 master's degree and PhD in Microbiology and Immunology from the University of Louisville, Louisville, KY, USA (2005-2008). In 2009, Dr AL Khodor worked as a postdoctoral fellow in the Signaling Systems Unit, laboratory of Systems Biology, at the National Institute of Allergy and infectious Diseases (NIAID), National Institutes of Health (NIH) in Maryland, USA. In January 2015, Dr Al Khodor joined the research department at Sidra Medicine where she acts as a Principal Investigator and was appointed as the Director of the Maternal and Child Health Department in July 2019.

A woman's health at conception and during pregnancy impacts the wellbeing of her child. Sidra's Maternal and Child Health (MCH) department aims to improve women's and children's health by implementing state-of-the-art clinical and translational research. This department focuses on two of the seven priority populations described in the Qatar National Health Strategy. In collaboration with the Obstetrics and Pediatrics clinics, the MCH department aims to address major health problems facing women trying to become pregnant, pregnant women, their growing fetus, infants and children up to two years old.

The goal of the department is to employ a systems biology approach by combining various omics tools (metagenomics, metatranscriptomics, proteomics, metabolomics, etc.) and non-omics tools such as dietary and lifestyle assessment in order to achieve an integrative view of health and identify signatures associated with disease.

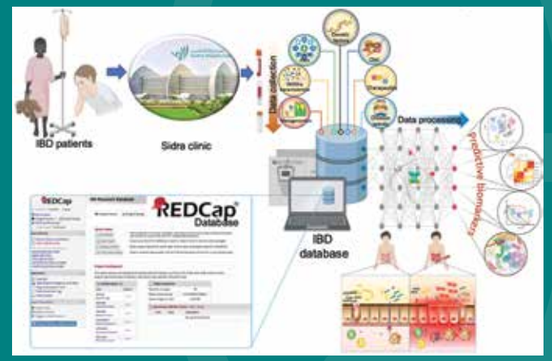
The Maternal and Child Health department focuses on the following areas: Pre-conception health, Prenatal health: healthy pregnancy leading to healthy babies, Identify novel biomarkers to predict pregnancy complications, Maternal Mental Health, Maternal Immune State and its impact on the Child's Health and Development, Baby's health: the first 1000 days.

Maternal and Child Health Highlight

Pediatric IBD Database: a Valuable Tool to Implement Precision Medicine in IBD

Inflammatory Bowel disease (IBD) is one of the conditions focused on by the Maternal and Child Health Department, directed by Dr. Souhaila Al Khodor. IBD is a chronic inflammation of the gastrointestinal tract of which there are two major types: ulcerative colitis and Crohn's disease. The phenotypic spectrum of children with IBD is highly variable, and studies describing IBD in children living in Qatar are still missing. While the incidence of IBD in children appear to be on the rise, it becomes vital to establish a national pediatric IBD registry in order to provide a comprehensive care of pediatric IBD patients in Qatar. It is also necessary to employ the latest technologies to dissect various pathophysiological profiles associated with IBD in the least invasive way and to offer patients with IBD the best treatment in a personalized fashion.

As part of the continuous efforts in our IBD study funded by QNRF: "Towards a systems-level understanding of pediatric Inflammatory Bowel Disease in Qatar: Interplay between the immunogenome and the microbiome" and in collaboration with Sidra Gastroenterology clinic, investigators at the MCH department has established the first Pediatric IBD database.



This database enrols all IBD patients that consent to be part of the study and provide a comprehensive assessment of their clinical status and diagnosis on regular basis. It does not only host phenotypic or clinical data from those patients, but will also include information about their genome sequence (if clinically indicated), their microbiome (microbes living in/on the human body) and their immune status in addition to other markers that are considered vital to differentiate between the disease subtypes. The database also includes information about the disease index, an important score to determine whether a patient is in relapse or remission, as well as previous and current treatments. Furthermore, this database is a step forward to implementing precision medicine in IBD patients as it enables the clinician to monitor specific microbial or immune signatures for the IBD patient during the follow up visits and can help predict their disease relapse/progression. It is also a great tool to assess a personalized response to a specific treatment.

Highly aligned with the National Priorities, the database is considered a vital support tool for identifying "Predictive and Prognostic biomarkers in IBD". Building and continuously updating this tool that hosts large datasets will be appealing for pharmaceutical and commercial companies to invest in more research in pediatric IBD.

A mother of a Qatari IBD patient enrolled in our study said: "We are very happy to be part of this study and to have this resource. We are hopeful that the team can find interesting results that can help improve patient's care".

Research Without Prior Consent: CONNECT 2

Dr. Colin Powell
Senior Attending Physician
Emergency Medicine and
Honorary Professor of
Child Health

Conducting research in Paediatric Emergency Medicine has unique challenges. How should we approach obtaining informed consent when researching into treatment for acute emergencies such as seizures, asthma attack or sepsis? In Europe and Australia, Research without Prior Consent, has been established as acceptable practice for recruiting children into studies of emergency clinical presentations. This is supported by extensive research with families and clinical staff illustrating that it is considered acceptable and ethical. In the USA there is the informed consent waver policy for emergency recruitment and procedures. Currently in Qatar we have no such legislation. We do not have a current understanding of views of families about recruitment to studies using this approach.

CONNECT 2 is a study examining the attitudes, acceptability and understanding of research, particularly focusing on acute situations. We will be interviewing families and older children, clinical staff about their views. The importance of this work is that it will establish the evidence base and justification for developing this approach to research in emergency situations in Qatar. In an acute life-threatening situation there is no time to obtain informed consent with families who will be distressed, and anxious about their child. Their child needs to be treated. We want to compare two acceptable treatments.

If a child fulfills the criteria for recruiting into the study, they can be randomized to either treatment arm. When the emergency is over, we go back to the parents to ask for consent. We want to find out whether this approach is acceptable in Qatar. This will truly give us a Qatari perspective. We have the opportunity to develop a precision medicine approach to managing acutely unwell children in conjunction with the genomic research laboratories in Sidra. Establishing this methodological approach to recruiting children to emergency studies will be the first step.

Clinical Research Interest Groups

Clinical **Research Interest Groups**

Clinical Research Interest Groups foster enhanced collaboration between clinicians and researchers. These groups align with the "One Sidra" philosophy on which the Precision Medicine Program is built. The purpose of interest groups is to coordinate research efforts within a specific field of clinical interest, and identify common questions that require multi-disciplinary approaches to answer.

01. I3 Immunity, Inflammation and Infection

The "Immunity-Inflammation-Infection" interest group focus on topics ranging from immunogenetics and tumor immunology to cellular/systems immunology. The projects undertaken by members of this group rely on a wide array of technologies that have been established in core facilities and individual laboratories that may be leveraged for the development of immune profiling assays.

01.1 Nephrology Working Group

The I3 group also focuses their research in the field of Nephrology.

01.2 Microbes and Infectious Disease

The "Microbes and Infectious Disease" group assumes a broad array of research topics in microbiology and infectious diseases. These topics encompass the classic trio of infectious diseases, pathogen and environmental factors that contribute to the development of disease in the host. Research Projects investigate: multi-drug resistant pathogens, the role of the hospital environment in hospital-associated infections, and the utilization of metagenomic sequencing for pathogen detection in sterile site samples is being examined.

01.3 Atopic Diseases

Is a subgroup of I3 and focuses on research projects associated with neonates and children showing symptoms of allergic diseases including those with asthma, food allergies and/or dermatological phenotypes.

02. T.U.M.M.I.E.

The group "Towards Understanding Microbiome and Mucosal Immunity in Inflammatory Bowel Disorders and Enteropathies" (TUMMIE) focuses on three major areas: translational research, clinical research and basic laboratory science. The research will be centered on gastrointestinal disorders including inflammatory bowel disease (IBD), enteropathies, celiac disease and other autoimmune bowel diseases. The focus of research is on implementing precision medicine for these Gastro-Intestinal (GI) disorders, developing new diagnostic tools and clarifying disease mechanisms, pathophysiology and pharmacogenetics.

03. Mother and Child

The "Mother and Child" (MCH) group at Sidra aims to improve women's and children's health by implementing state-of-the-art clinical and translational research. This department focuses on three of the seven priority populations described in the Qatar National Health Strategy. This working group is comprised of members from different Sidra clinics such as Obstetrics, Psychiatry for Women›s Mental Health, Gynecology, Acute Care Medicine and Neonatology Clinical Services as well as regular members from Hamad Medical Corporation (HMC). MCH group discuses major health problems facing women trying to become pregnant, pregnant women, their growing fetus, infants and children up to two years old.

04. Neurology and Psychiatric Disorders

This is a multidisciplinary working group involving scientists and clinicians at Sidra, aiming to enable integrated, state-of-the-art translational research on paediatric diseases relating to Neurology, Psychiatry and Developmental disorders, which constitutes a sizeable health burden in Qatar and beyond. Members of this group use a variety of techniques ranging from computational such as bioinformatics and image processing to experimental methods such as genome sequencing, disease modelling and behaviour-based tools. Notably they have been developing tools to improve genetic testing, counselling as well as biomarker discovery. This group held regular meetings to discuss ongoing projects, ideas to cover existing gaps and improve standard care, as well maximum synergies and use of resources. As a major outcome, it has led to the identification of priority disease cohorts and venues for research capacity building though establishing a nationwide program as a key pillar to the Qatar Precision Medicine Institute as part of the precision medicine mission in Qatar.

05. Acute Pediatrics

Emergency and critical care are major paediatric specialties involving acutely unwell and injured children. The Acute Paediatrics Research group focuses on research into the best way to manage paediatric emergencies; in the Emergency Department, in the Paediatric Intensive Care Unit (PICU) and the inpatient wards. The clinical expertise in the Emergency Department is managing undifferentiated illness, resuscitation and instigation of life saving procedures with definitive treatment and managing the primary care secondary care interface safely and effectively. This research group will combine scientific expertise within the research laboratories with clinical translational expertise of the clinicians in emergency and critical care.

06. Cancer Precision Medicine

In the "Cancer Precision Medicine" group, there is a strong determination to understand the reasons for treatment failure, match each patient with the most appropriate treatment, develop novel therapeutic approaches with emphasis on targeted therapy and immunotherapy, identify the genetic basis of cancer risk and progression, and determine biomarkers of outcome and toxicity. The group is developed around the 3 Sidra Precision Medicine Pillars: Biorepository, Advanced Diagnostic, and Advanced Therapeutics, and in alignment with National Priorities. The group is constituted by researchers and clinicians working on cancer from a research and patients' service prospective.

07. Cardiac Working Group

The Cardiac group observes an extensive diversity of congenital disease with a personalized approach for identification of genetic variants and clarification of the pathways to find possibilities for new/improved treatment. In the Cardiac group, there is an interest to test and develop new cardiac devices which can improve patient health and treatment. Cardiology Fellows are also encouraged for active participation in such studies. Moreover, the group recognizes the importance of providing long-term interventional clinical studies to enable patients to enroll in the hope of improving the clinical outcome.

08. Diabetes Endocrinology and Metabolic Disorders

In the Diabetes, Endocrinology, and Metabolic Diseases working group, by recruiting patients suffering from obesity, diabetes and/or endocrine disorders, the group aims to develop a detailed map of the epidemiology of these diseases in Qatar. The stratification of the different types of these diseases, in combination with genetic approaches and functional studies using animal models, will allow researchers to implement personalized therapeutic strategies and interventions for children and other patients affected by specific illnesses.

09. Genome Data and Digital Health

In the Genome Data and Digital Health working group, questions such as how to best integrate genomics and genome sequencing in patient health are addressed. Large datasets of patient genomics data sequenced as part of different projects, are assessed by scientists using different applications. These applications include bioinformatics, genomic landscape analysis and phenotypic data integration. An important operation in the Genome Data and Digital Health working group is to pre-address database development and standardization addressing growing datasets in genomics, which can be used for both Sidra Medicine projects as well as by partners, such as the national Qatar Genome Programme (QGP). Furthermore, this group ensures genomic data standardization such as GA4GH is followed in order to work with the increasing Genomic data.

Implementation of Clinical Metagenomics-based Diagnostics for Infectious Diseases

Dr. Mohammad Rubayet Hasan

Clinical Molecular Microbiologist
Pathology Sciences

"Our study is aimed to improve diagnosis and management of patients with critical infectious diseases. Very few laboratories in the world have the in-house expertise and experience of offering NGS as a diagnostic service for infectious diseases. It is expected that once implemented at Sidra Medicine, NGS will help Microbiologists and Infectious Disease physicians to manage some of the most diagnostically challenging medical cases associated with complicated infections of vital human organs and body fluids. If successful, the technology can also be transferred to other health-care facilities in Qatar for implementation at the national level."

Infectious diseases account for 8% of all deaths in Qatar. Infectious diseases pose a significant challenge to the public health system of the country. Sidra Pathology aims to improve diagnosis and management of patients with critical infectious diseases. Over the years, Sidra Pathology has accelerated their research contribution in the field of laboratory medicine. They pioneer in applying novel molecular technologies to the diagnosis of infectious diseases along with significant ongoing efforts to evaluate and implement new commercial and in-house developed tests for infectious diseases.

With both Sidra and QNRF funding, Mohammad Rubayet Hasan, a Clinical Molecular Microbiologist, and his team have focused their research in establishing a standardized Next Generation Sequencing (NGS) based clinical metagenomics approach and implement the test on a pilot basis for patient services at Sidra Medicine. The clinical metagenomics approach is built on the principle of 'precision medicine', in which all nucleic acids present in clinical samples are sequenced to detect and identify pathogens and infer their antibiotic resistance pattern. The ongoing research has shown promising results where NGS has been successfully applied to detect pathogens in cerebrospinal fluid specimens with >95% sensitivity and specificity. The NGS is a powerful technology in the diagnostic field but its role in infectious disease diagnostics is still very limited. The researchers working on this project are confident that the application of NGS will particularly benefit patients with invasive, sterile body site infections, which remain undiagnosed by culture and standard molecular test methods.

Custom Valves: A Precision Medicine Approach to Congenital Heart Defects

Dr. Ziyad M. Hijazi

Acting Chief Medical Officer &
Chair of the Department of
Pediatrics

"Clinical research is a key part of our remit at Sidra. As surgeons and physicians we are committed to ensuring that we never stop researching new treatment and technologies that can save lives and ease the care and treatment of our patients."

Congenital Heart Defects are the most common type of birth defects (approximately 1% of all births result in congenital heart disease) that are the leading cause of birth defect-associated infant illness and death. Serious congenital heart defects typically require open heart surgery or other invasive procedures within the first year of life, and is followed up by other surgeries and procedures into adulthood.

Tetralogy of Fallot, a common congenital cardiac defect. Traditionally, patients with this defect are sent for open-heart surgery within the first few months of life. Many of these patients end requiring a valve to be inserted between the right ventricle and pulmonary artery. Some of these patients can benefit from a minimally invasive procedure to place an artificial valve which is administered through the groin region. However, majority of the patients cannot benefit from this minimally invasive procedure. Almost 80 per cent of patients with this defect have to undergo open-heart surgery because the native area of the valve is larger than the approved available artificial valve diameters.

Dr. Hijazi, an interventional cardiologist is working to implement minimally invasive procedures for such patients, by practicing Precision Medicine which would help drive Sidra Medicine's clinical research agenda. With his expertise, Dr. Hijazi, assigned as a Global Principal Investigator in collaboration with a Chinese Medical Device Manufacturer that developed and fashioned a valve that would benefit patients who were previously bound to undergo an open-heart surgery. Once such patient of Dr. Hijazi came seeking help from Dubai, for whom administering the approved valve was highly unlikely due to a larger diameter.

Dr. Hijazi in collaboration with the manufacturer, successfully fashioned her treatment plan and ordered a custom-made artificial valve based on a 3D model of her heart. The patient successfully underwent placement of the valve in her pulmonic position and has safely gone on to deliver healthy twin babies.

Diabetes and Obesity Research at Sidra Medicine

Prof. Khalid Hussain
Division Chief of Paediatric Endocrinology
Chair of Medical Education

The research in the Division of Paediatric Endocrinology focuses on understanding the causes of diabetes mellitus and early onset obesity in children as well as developing new technologies in managing childhood diabetes. Research in childhood diabetes and obesity is a national priority area for the State of Qatar and thus our research fits very well with national priorities. Over the last 2 years we have identified every child (from birth to 18 years) with diabetes in the State of Qatar and begun to unravel the biochemical and genetic mechanisms that lead to diabetes in all these children. We now know the causes of diabetes in every child and have started implementing treatments based on the underlying biochemical and genetic mechanisms thus bringing precision medicine into the clinical setting.

Our research has shown that Qatar has the 4th highest incidence of childhood type 1 diabetes mellitus in the world. Some of these children with type 1 diabetes are now managed with cutting edge technology (using glucose sensors and insulin pumps) with Sidra Medicine being a world leader in this technology. This work is driven by my colleague Dr. Goran Petrovski.

Children with type 2 diabetes are now being treated with a new medication (Liraglutide) which is expected to help with weight loss and control of their blood glucose. In addition, in some patients we have been able to stop the daily insulin injections and put these patients onto oral medications. This has been completely transformational for these patients. In our obesity research we have identified children with rare genetic causes of early onset obesity.

As an example of precision medicine in this area, a Qatari child presented with severe obesity (28kg at 18 months) where we found the genetic cause of the obesity and she has now been treated with a medicine tailored to the genetic defect. Her weight is now 15kg after 1 year of treatment. Her parents are delighted with the transformation in her life. She is now a completely normal healthy child.

Research Principal Investigators

Research **Principal Investigators**

Ammira Akil, EMBA, PhD, Diabetes Laboratory

Dr. Ammira has a bachelor's degree in veterinary medicine and surgery, MSc in molecular Immunology, MSc and graduate certificate in university teaching and learning, PhD in molecular genetics from university of New South Wales, Australia and Executive MBA from HEC Paris business school in Qatar. Dr. Ammira research focus on the molecular genetics of diabetes mellitus, in particular, her interest focuses on diabetes complications prevention and precision diagnosis. During her career, Dr. Akil was a finalist at the Inventor of the Year Award and filed one Australian provisional patent application with the New South Innovations, Australia. She has also received several prestigious national and international recognition awards.

Annalisa Terranegra, PhD, Laboratory of Nutritional Genomics and Metabolism

Dr. Terranegra, PhD in Molecular Medicine and Nutritional Sciences, joined Sidra Medicine in 2014, where established the Laboratory of Precision Nutrition interested in the effect of diet on gut microbiota and epigenetic mechanisms. In collaboration with local and international institutes, the Dr. Terranegra's lab runs studies on diabetes, obesity, cardiovascular diseases and perturbation of the mother-baby axis. Dr. Terranegra covers also teaching roles as adjunct assistant Professor at University of Milan, Italy (2007-2013), Hamad bin Khalifa University (2015-2020) and Qatar University, Qatar (2018-present).

Aouatef Ismail Chouchane, MD, Laboratory of Dermatology

Dr. Ismail Chouchane received her MD degree from the Faculty of Medicine of Sousse, Tunisia. Following four-year residency training in Dermatology & Venereology at several teaching hospitals in Tunisia and France, she obtained her Medical Specialty Degree in Dermatology & Venereology. She then joined the US National Institute of Heath for a fellowship in biomedical research during which she worked on several areas of Dermatology Research. With her more than 24-year clinical experience, she held the positions of Consultant and Senior Consultant in Dermatology and Venereology at several medical institutions. Throughout her career, besides her work as clinician, Dr. Ismail Chouchane held research positions among which a position at Weill Cornell Medicine-Qatar, prior to joining Sidra Medicine as a principal investigator where she built the first Dermatology Research group in Qatar.

Research **Principal Investigators**

Bernice Lo, PhD, Regulation and Tolerance Laboratory

Dr. Lo is a Principal Investigator in the Human Genetics Department at Sidra Medicine. Dr. Lo performed her post-doctoral training in the Laboratory of Immunology at the National Institutes of Health in the US. She is trained in cell and molecular biology and genomic approaches for genetic diagnosis. During her fellowship, she helped discover and understand the etiology of two new diseases of immune dysregulation. She received her Ph.D. in Cell Biology at Duke University, where she began her appreciation for the immune system and the critical role of immune tolerance and regulation.

Cristina Maccalli, PhD, Advanced Cell Therapy Core

Dr. Maccalli is a Principal Investigator at the Advanced Cell Therapy Core. Her expertise is in the field of immunology, tumor immunology and immunotherapy. She has carried out her post-doctoral research programs at the National Cancer Institute in Milan, Italy and, then, as visiting fellow at the Surgery Branch, National Cancer Institute, NIH, Bethesda, MD, USA. Prior to joining Sidra Medicine, Dr. Maccalli has contributed to novel studies aimed at the immunological characterization of cancer stem cells, the development of Phase I/II immunotherapy clinical studies and immunomonitoring of cancer patients during her time as a senior investigation at the San Raffaele Foundation Scientific Institute, Italy. She has also contributed to the development of the Laboratory of the Italian Network of Biotherapy of Tumors (NIBIT)/University Hospital of Siena, Italy dedicated to ImmunOncology (IO) studies and the immunomonitoring of patients undergoing immunotherapy treatments.

Luis R. Saraiva, PhD, Laboratory of Neurogenetics, Behavior and Disease

He completed a Licenciatura in Biology at the University of Evora (Portugal). After, he became a Fellow of the International Graduate School in Genetics and Functional Genomics of the University of Cologne (Germany), where he received his PhD in Genetics. After a brief period as a visiting scientist at Harvard Medical School in Boston (USA), he worked as a post-doctoral scholar in the lab of Linda Buck (Nobel Laureate in Physiology and Medicine 2004) at the Fred Hutchinson Cancer Research Center in Seattle (USA). As he became an EBI–Sanger Postdoctoral (ESPOD) Fellow, he moved to Cambridge (UK), where he continued his postdoctoral training at the EMBL-EBI and the Wellcome Sanger Institute. Since October 2015, he is a Principal Investigator and Director of the Metabolism and Diabetes Program at Sidra Medicine.

Research **Principal Investigators**

Mohammad Haris, PhD, Molecular Imaging Laboratory

Dr. Haris received his bachelor's degree in life sciences with major in Chemistry, and later completed his master in Biochemistry. He earned his Ph.D. in Biomedical Imaging from Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow, India. Dr. Haris went on to do his postdoctoral fellowship in the department of Radiology at Perelman School of Medicine at the University of Pennsylvania, Philadelphia, USA, and subsequently became research scientist in the same department. During his tenure at Pennsylvania, he developed various novel MR imaging methods to image metabolites in vivo with potential applications in cancer, cardiovascular disorders, and neurological & neurodegenerative diseases. He was the first to develop an MR method for imaging cellular energetics by targeting creatine recovery kinetics using CEST MRI. Dr. Haris joined Sidra Medicine as a Principal Investigator in October 2013.

Nicholas Van Panhuys, PhD, Asthma and Allergy Laboratory

Dr. Van Panhuys completed his BSc in Biochemistry and Molecular Biology, Cell and Developmental Biology at Victoria University (New Zealand). Following this, he completed the Honors program in Molecular Biosciences at Victoria University. Before being awarded Rex and Betty Coker Post Graduate Scholarship to conduct his PhD studies at the Malaghan Institute for Medical Research (New Zealand) where he investigated the role of IL4 and STAT6 in protective immunity and T helper 2 immune responses. Consequently, he was appointed as a research fellow in the Laboratory of Systems Biology at the NIAD, NIH. Since July 2015, he has worked as a Principal Investigator and head of the Laboratory for Asthma and Allergic Disease at Sidra Medicine.

Nico Marr, PhD, Systems Immunology and Immune Deficiency Laboratory

Nico Marr received a University Diploma in Biology and a Doctor of Science degree from the Julius Maximilian University of Würzburg, Germany, for studies in the field of Microbiology and Immunology. During his doctoral and early postdoctoral research, which he conducted in the Department of Microbiology & Immunology at the University of British Columbia, his research was focused on host-pathogen interactions and immune evasion mechanisms of Bordetella pertussis, the etiological agent of whooping cough. Later on, he completed a Postdoctoral Fellowship in Respiratory Health and worked as a Research Associate at the British Columbia Children›s Hospital Research Institute in Vancouver as well as at the Canadian Center for Vaccinology located in the IWK Health Centre in Halifax, Canada. During that time, his studies focused mainly on the ontogeny of the human immune system and immune defenses in early life against another important and common respiratory pathogen, human respiratory syncytial virus (RSV). Since 2015, he is an Investigator and Research Group Leader at Sidra Medicine.

Research **Principal Investigators**

Sara Deola, MD PhD, Advanced Cell Therapy Core

Dr. Deola is a clinical scientist with a Board Certification in Hematology-Oncology from University San Raffaele Vita-Salute, Milan, Italy and a PhD in Experimental Hematology from the University of Milan Bicocca, Italy (training at Telethon Institute for GenE Therapy, TIGET). She completed a post-doc fellowship at NIH, Bethesda USA at the Clinical Center, Department of Transfusion Medicine, Section of Immunogenetics. She joined Sidra Medicine in 2014, as Bone Marrow Transplant Program Manager in Translational Medicine/Research, and is holding a PI position since June 2017.

Wouter Hendrickx, PhD Functional Cancer Omics Laboratory

Dr. Wouter Hendrickx is an investigator in the immunology, Inflammation and metabolism department and member of the Cancer Precision Medicine working group at Sidra Medicine. He is the PI of the Functional Cancer Omics lab and has experience in stem cell and cancer research at the universities of Brussels (VUB), Leuven (KUL) and Norwich (UEA). Where he gained an MSc in biomedical Science (2004) and an MSc Bio-informatics (2005) and a PhD in Medical Science respectively (2012). He has worked on several different projects relating to the tumor micro environment including extensive work on the role of MMP's and the degradome. He has experience with classic molecular biology techniques as well as advanced 3D cell culture and proteomics technology. At Sidra he has focused since 2014 on the tumor immune micro environment deploying bio-informatic tools to analyze gene-expression data form bulk tumor for immune related signatures and other determinants of the immune phenotype and translating the findings to the wet lab environment. Since 2019 he leads Sidra Medicine's efforts in establishing a Biorepository for Pediatric Cancer Patients.

Younes Mokrab, PhD, Medical and Population Genomics Lab

Dr. Younes Mokrab is principal investigator and head of Medical and Population Genomics Lab, Sidra Medicine. He joined in 2015 from Eli Lilly, where he led computational genomics research at the Neurogenetics Discovery Unit to identify/validate drug targets and stratify patients with neuropsychiatric disorders including Schizophrenia, ALS and Parkinson's, working closely with Psychiatric Genomics Consortium (PGC). Dr Mokrab obtained a BSc in Genetics from the University of Sheffield, UK (2003), a PhD in Bioinformatics University of Cambridge, followed by a postdoctoral fellowship from University of Oxford. Next, he moved to the Pharmaceutical industry, initially Lonza Biologics performing in silico bio-pharmaceuticals engineering and subsequently Eli Lilly where he led research in early-stage drug discovery in various areas of neuropsychiatry. Upon joining Sidra, Dr Mokrab helped establish research programs in population and medical genetics and is a co-founding member of the Qatar Genome Programme Research Consortium.

Approached Respiratory Diseases with Precision Medicine

Prof. Ibrahim Janahi
Division Chief of Pulmonology
Chair of Medical Education

"As a clinician scientist, I practice evidence-based medicine which is the source of my ideas and research questions. My research is designed to answer questions that arise during clinical practice."

Medical Research explores better ways of diagnosing, treating and caring for patients. Clinicians are central to the journey of a research study. Clinicians bridge the work done by other researchers, biostatisticians, epidemiologists by translating research into clinical practice with patients. As a pulmonologist, Professor Ibrahim Janahi closely studies genetic bases for chronic lung disease like Cystic Fibrosis, Asthma, Chronic Bronchitis, and others. Prof. Janahi uses his clinical practice to understand ailments, symptoms and signs to make sense of different factors that end up showing as a result of a disease or a syndrome.

One such case of research impacting patients was the identification of genetic defects causing cystic fibrosis having mutations that are specific to the Qatari population. This resulted from decades of research done by Prof. Janahi and his team. Now, his research focuses on the implications of these mutations. Through extensive research and drug testing, it was identified that that the cystic fibrosis mutation amongst the Qatari population has two different classes of genetic mutations. In collaboration with researchers across the world in north America and Europe, the Qatari genetic mutation is further being studied and tested to better administer drugs that result in higher efficacy amongst Qatari's with Cystic Fibrosis.

Dr. Janahi also conducts research focusing on the association between asthma and obesity amongst children. Due to extensive studies Prof. Janahi and his team have found that obese asthma children do not respond as effectively to treatment as a lean asthmatic child. As a clinician scientist, Prof. Janahi and his team started to explore mechanisms behind response to medications of Obese Asthmatic children.

Animal research has been crucial in the development of this type of research. During initial stages, it was found that the airway muscles of obese and lean mice are significantly different, thus confirming that obese mice have asthma symptoms due to a twitching activity in their airway muscles. An imbalance in sphingolipid (type of lipids) pathways is proposed as a mechanism to explain airways hyperresponsiveness without inflammation in obese individuals with asthma. Prof. Janahi hopes this breakthrough will help administer better treatment for obese asthmatic children in Qatar by identifying new target pathways.

Core Services

- Digital Health
- Integrated Genomics Services
- Deep Phenotyping Core
- Advanced Cell Therapy Core
- Research Operations and Services
- Biostatistics Core
- Clinical Trials Office

Core **Services**

Digital Health

The Digital Health Core is led by Dr. Rashid Al Ali. Before joining Sidra Medicine, Dr. Rashid Al Ali was the Director – Corporate Services at ASPIRE Academy for Sports Excellence. He was responsible for overseeing the operations of Information Technology, Finance & Procurement, Human Resources, Communication and General Administration. At ASPIRE, Dr. Rashid Al-Ali worked closely with Senior Management and External Consultants on developing the Corporate Vision, Mission and Strategy map based on the balance score card system. Dr. Rashid Al-Ali received his Ph.D. in Computer Science from Cardiff University - Wales, UK, and his MS in Computer Science from George Washington University - Washington, DC, USA. Furthermore, Dr. Rashid Al-Ali graduated with a BS in Computer Engineering from the University of the Pacific - California, USA.

Dr. Rashid Al Ali
Division Chief,
Bioinformatics – Digital Health

The emergence of fast and efficient technologies for the sequencing of nucleic acids and proteins, pushes Bioinformatics to produce an ever-increasing amount of experimental data along with technological advances and the ubiquity of the Internet that gives scientists unparalleled opportunities to access, share and analyze vital data and information stored in many datasets. The digital health department offers high-quality services to all clinical and research initiatives to assist clinical and research scientists by providing bioinformatics, computing and storage resources, helping them understand and manage their datasets.

The department aims at creating a digital ecosystem to support all computational aspects (compute, storage, software development, data processing, data management & analytics) that are crucial for advancing precision medicine within Sidra and Beyond. It also provides solutions for customized scientific tools and web-based research data analytics and visualization portals for communicating scientific results of the underlying research data as an integral part of the scientific process. Thus, unifying various aspects of biomedical, computational and precision medicine research.

Bioinformatics Highlight

Digital Health at Sidra Medicine

The different teams in the Department of Digital Health work together to offer Sidra researchers unique and customized informatics services. All work is executed either entirely by custom-design or based on existing industry tools, built in the Sidra research department for specific requirements of multiple research groups.

Precision Medicine Experience
Informatics Platform
Sidra Pediatric Research database

Sidra Pediatric Cancer Registry: This Registry is a modern web application developed from the ground up to be the main facilitator of all cancer research involving the patient population present at Sidra Medicine. This resource is central to Sidra Medicine›s current and future cancer research efforts. The registry established during this project also serves as a reporting tool for the Qatar Cancer Registry mandated by MOPH. This web application is currently being used by the Functional Cancer Omics Laboratory.

Sample Inventory: The Sample Inventory web application provides the functionality of the Bio Repository for Sidra Medicine Researchers, which is used to store information on bio-specimens to support future scientific investigations. Currently, the following features are supported: (i) Users are created and provided access to their respective projects; (ii) Family details of samples may be entered and maintained; (iii) Collection of samples and details may be entered and maintained; (iv) Summary statistics of samples and associated details for each sample inventory may be viewed.

This application is currently being utilized by the Human Genetics Group.

REDCap for PMO: The Research Project Management Office (PMO) has started using REDCap this year to improve the way research projects and related documents are organized.

The Digital Health Division is continually working behind the scenes to provide support for technical and back-end programming to provide the requested and enhanced functionality for PMO REDCap. Working collaboratively with the PMO Office, it was possible to improve existing processes and workflows by implementing an electronic system to enable automatic reminders of IBC, IRB, etc. of the expiry notification to the research staff.

cBioPortal FOR CANCER GENOMICS

cBioPortal: The cBioPortal is an open-source resource for the interactive analysis of multidimensional cancer genomics data sets. The Digital Health Division locally hosted the CBioPortal app and loaded the required data for the TCGA study, the Pediatric Public Studies, and the Sidra Colon Cancer Specific Study to empower researchers to translate these rich data sets into biologic insights and clinical applications. This application is currently being used by the Cancer Immunogenetics Laboratory.

Gene Retriever: Researchers in the life sciences research community often ask, «What genes are referred to in the results of my PubMed search? «Gene Retriever, a collaboration between Acumenta Biotech and Sidra Medicine, answers this critical question by drawing up gene lists from the results of any PubMed search. Implementation of the Gene Retriever software by the Digital Health Division at Sidra uses multi-threaded programming techniques to search in parallel for more than 20 million abstracts and produces results in less than 2 minutes, which is an improvement of approximately 60 times faster than the time taken by the Acumenta LitLab software previously used for the same purpose.

The application is currently being used by the Translational Systems Biology Laboratory team.

Core **Services**

Integrated Genomics Services

Clinical Genomics Lab, Omics, Applied Bioinformatics and Zebrafish Core

The Integrated Genomics Services Core is led by Dr. Stephan Lorenz. Dr. 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. 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 focused on high-throughput single-cell genome and transcriptome sequencing. He and his team developed methods to rapidly process thousands of cells per experiment using high-end automation to achieve high throughput and to minimize assay volumes and cost.

Dr. Stephan Lorenz
Director,
Integrated Genomics Services

The Clinical Genomics Lab, Omics, Genomic Data Science, and Zebrafish Core Facilities deliver genomics, molecular biology, and informatics services to researchers across Qatar. The Facilities aim to provide high-quality service and data i) by ensuring adherence to validated standard procedures, ii) by ensuring sample integrity and traceability iii) in a timely fashion with iv) excellent communication throughout a project's lifecycle. The unique combination of laboratory and analysis services allows IGS to deliver complex scientific projects from the initial screening of large cohorts to follow-up validations using targeted assays to data analysis and experimental follow-up in model systems. The routine offering of medium- and high-throughput sequencing services are enhanced by 3rd generation genome and transcriptome analysis methods and single-cell services in close collaboration with our Deep Phenotyping Core. Beyond the routine services, all groups are experienced in method development and always excited to work with our users to deliver novel and innovative approaches to genomics in research and healthcare.

Integrated Genomics Services Highlight

High-Throughput Genomics Paves the Way for Genetics-Driven Diagnostics and Care in Qatar

Since its inception, Sidra has strived to use the power of genomics for the benefits of patients and researchers in the whole country and beyond.
To achieve this goal, Sidra has established and now operates the largest and most advanced infrastructure for genomic sequencing and analysis in the country, in a collaborative effort across multiple departments.

Sidra's Integrated Genomics Services department is operating laboratory pipelines to isolate, prepare and sequence genetic material from a wide range of biological sources using automated instruments and high-throughput sequencers with an annual capacity of up to 20,000 samples. This effort, led by a dedicated team of scientists, generates petabytes of data, which is processed and stored on Sidra's high-performance computer. The Genomic Data Science group (GDS) of the IGS department implements robust, high-quality pipelines that allow the processing and primary analysis of genomic data in a matter of hours for research and clinical use. Sidra's Biomedical Informatics (BMI) department provides invaluable support for Sidra's projects by maintaining and expanding the compute infrastructure and data storage systems and by distributing these big datasets to researchers across the globe, allowing them to use this data for high impact discovery. Further, BMI is leading several efforts to apply Artificial Intelligence and Machine Learning to genomic data and extract novel insights relevant to population health.

The data generated and analysed so far covers over 23.000 whole genomes and 20.000 transcriptomes and has been used for both internal and external projects, including international collaborations and clinical trials.

Leading the effort to implement Precision Medicine on a national scale, Sidra is applying insights gained from its research to support the diagnosis of Sidra patients with suspected genetic disorders and to study the genetic architecture of the local population at very high depth. This will lead to the identification of the underlying genetic causes of frequent disorders, thus closing the loop from research to the translation of science into clinical care and diagnostics.

Core **Services**

Deep Phenotyping Core

Dr. Jean-Charles Grivel
Director,
Deep Phenotyping Core

The Deep Phenotyping Core is led Dr Jean-Charles Grivel. Dr. Grivel obtained his Ph.D. in immunology from the University of Aix Marseille II. He was a postdoctoral fellow at the National Cancer Institute (NIH, USA) and became a Staff Scientist in 2000 at the National Institute of Child Health and Human Development. He pioneered the development of human organ culture for studying the pathogenesis of HIV, Human Herpes Viruses and Measles virus as well as their interactions, which led to the granting of two patents. Dr Grivel has developed several high dimension immunoassays for the quantification of viral antigens and other immune system related proteins. He has also developed several flow cytometric methods for characterizing antigen-specific cellular responses as well as submicron particles. These methods are used for studying the role of microvesicles in heath and disease, especially in cardiovascular disease and cancer as well as to characterize the antigenic composition and maturation of viral particles. Dr Grivel has received the NIH Award of Merit in 2006. He currently directs the Deep Phenotyping Core of Sidra Medicine.

Sidra Medicine is aiming at delivering personalized medicine to the patients of Qatar and beyond. The systematic measurement and analysis of qualitative and quantitative traits of patients, known as Phenomics, completes the personalized medicine approach initiated by genomic approaches. The Deep Phenotypic Core provides a multifaceted phenomics platform dedicated to establishing cellular, molecular and functional phenotypes that complement genomics, transcriptomics, and clinical phenotypic analyses of patients. Relying on metabolomics, lipidomics, elemental chemical analysis, high-dimension proteomics, super-resolution microscopy and high-dimension flow cytometry, the DPC generates systematic, high-quality, validated precise molecular and cellular phenotypes of patients and enable true phenomic science in Qatar. The DPC aims at providing in depth and breadth the panels of diagnostic and investigational assays proposed in Sidra and in Qatar.

The DPC mission is to provide the technical and intellectual frameworks for the realization of the second pillar of Sidra Research Personalized Medicine agenda: “Establishing an Advanced Diagnostics program”.

Deep Phenotyping Core Highlight

Serelogical Testing Methods for Better COVID-19 Tests

Sidra Medicine is supporting the need for better COVID-19 tests by developing alternative serological testing methods by working on establishing Immunity Passports, which have a “risk-free certificate” that would enable individuals to travel or to return to work with the assurance that they are protected against re-infection. Serological testing looks for the presence of antibodies produced by the immune system in response to an infection. It is an important tool for monitoring infectious diseases as it can detect past exposure to pathogens and, in certain cases, it can predict resistance to further infection if the immune response is protective.
Serological testing looks for the presence of antibodies produced by the immune system in response to an infection.
This response often outlasts the presence and therefore the ability to detect the infectious agent that caused this response.
Therefore, serological testing is an important tool for monitoring infectious diseases as it can detect past exposure to pathogens and in certain cases predicts resistance to further infection if the immune response is protective (as it is the case for most vaccines).
In the case of COVID-19, most serological assays use rapid tests that detect antibodies against a protein unique to SARS-CoV-2, the virus causing COVID-19 or

to a protein common to many Human Corona Viruses (hCoVs). The Deep Phenotyping Core team, in collaboration with Sidra Medicine’s Pathology Department has developed a serological assay that detects the immune response to proteins specific to SARS-CoV-2 as well to the 5 other hCoVs resulting in a more sensitive and specific measurement of the immune response to SARS-Cov2, by avoiding the false positive problems due to the immunity against other hCoVs, that have crippled other assays. In addition, Sidra’s assay measures many more types of antibody responses than current assays, allowing for the identification of truly protective responses, which are the basis for the establishment of an immunity passport and for the identification of potential donors of hyperimmune plasma for serotherapy. In particular, Sidra’s assay is different from other commercial assays because it measures the presence of the type of antibodies that are involved in preventing infections in mucosa surfaces from which the virus gains entry into the patient airways. The assay developed is a high-throughput assay allowing at the moment the measurement of up to 1000 samples daily, at a low cost.
Laboratory testing for the SARS Coronavirus-2 (SARS-CoV-2) has been one of the most important tools for controlling the COVID-19 pandemic. Not only has it been important to find out which people currently have the virus, but it is also important to know which people have recovered from the virus. The performance of commercially available tests for detecting antibodies against SARS-CoV-2 is highly variable and has hampered the development of immunity passports.
The preliminary results show that the Sidra Medicine test is much more accurate than current available antibody tests and provides more nuanced information about the immune response. The teams are looking forward to clinically validating this assay and to better define the correlates of protection against COVID-19.

Core **Services**

Advanced Cell Therapy Core

The Advanced Cell Therapy Core (ACTC) is led by Dr. Chiara Cugno. Dr. Cugno is an experienced medical doctor with a Board Certification in Pediatrics and Pediatric Hematology/ Oncology at the University of Pavia, Italy, and a II Level Master in Pediatric Hematology at the University "La Sapienza" of Rome, Italy. Dr. Cugno joined Sidra Medicine in April 2014, and has been working on the development of the Advanced Cell Therapy Core, including a Cellular Therapy Unit for the delivery of cellular products for tissue, cell and gene therapy, and research projects on Mesenchymal Stromal Cells and pediatric leukemia

Dr. Chiara Cugno
Director,
Advanced Cell Therapy Core

The Advanced Cell Therapy Core (ACTC) synergistically completes Sidra's Precision Medicine Program to provide personalized treatments to patients. The Advanced Cell Therapy Core (ACTC) is designed to provide advanced personalized care in a patient's path from diagnosis to intervention by tailoring treatment options in the fields of:

Hematopoietic Stem Cell (HSC) Transplantation, e.g. processing of HSC; Cell Therapy, e.g. production of third party Mesenchymal Stromal Cells (MSC) as off-the-shelf treatment for several clinical applications; Regenerative Medicine, e.g. production of platelets derivatives; Gene Therapy, e.g. replacement of a faulty gene.

These treatments promise to reverse often debilitating manifestations of diseases such as cancer, genetic and metabolic disorders that disproportionately affect children, as well as provide opportunities for therapies using stem cells for rejuvenation and regeneration in pediatric and maternal care.

Core **Services**

Research Operations and Services

The Research Operations and Services Core is led by Max Renault. Mr. Renault has a background in Engineering and has extensive expertise in Project/Program Management, R&D, New Product Development, Operations Management, Business Development and Technology Commercialization. Mr. Renault has worked in Europe, Far-East and Middle-East in the fields of Telecommunications, Manufacturing, Aerospace/Defense, Formula One and Biomedical Research. He is currently a doctoral candidate for a PhD in Innovation Management.

Max Renault
Director,
Research Operations and Services

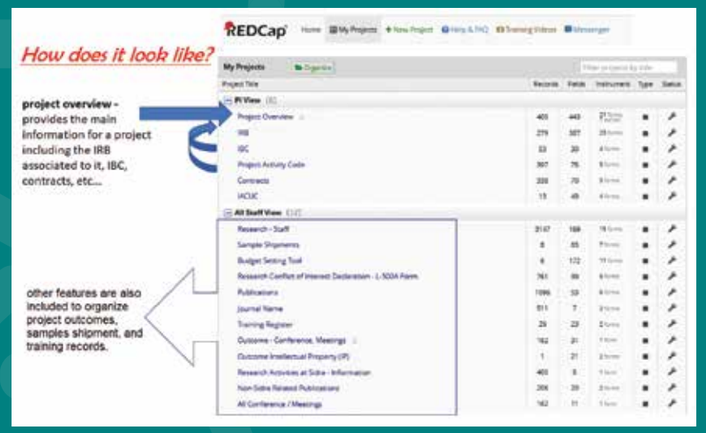
Our mission is to offer a "one-stop-shop" approach and solution-oriented admin service to researchers and clinicians. We help establish good laboratory practice, agile and fit-for-purpose processes, and high standards for managing research in a compliant manner. Our main administrative functions are as follows. The Project Management Office serves as a central repository and enabling service for research studies. The Laboratories & Biosafety Office ensure labs are in good working condition and practices safe. The Grants Office manages external awards. The Business Office handles budgets, financial reporting and procurement matters. The Outcomes & Reporting Office looks after strategic reporting, communications and outreach. Finally, the Research Contracts Office manages collaborations and other research agreements. As a group we are the primary interface and facilitators between scientists and internal/external parties, and lead commercialization of research services.

Research Operations and Services Highlight

REDCap for Project Management

In 2020, the Research Project Management Office (PMO) established a central database and system to manage the lifecycle of research projects. With the support of our Advanced Application Team, the solution was based on REDCap and has been fully developed within Sidra Medicine at no cost. It was a big step away from multiple and separate excel sheets into a new central intuitive database, integrating all relevant project information and documents in one place. The focus lay on creating a common system that brings Research administrators and researchers together, without introducing additional burden.

As a project holder, it is very difficult to keep a "live" updated overview of all project documents and compliance to relevant national regulations and Sidra policies. We can now view the most important information (e.g. reports due, IRB or IBC expiry dates, etc.) in a single dashboard. The database allows the PMO to generate reports and analyze gaps, which are then fed back to the project holder. This makes the identification of missing documents easy, e.g. a declaration of interest for a team member. In addition, the PMO REDCap was developed to capture every aspect of a project and its associations. For instance, with a single click we can view the project's regulatory approvals (e.g. IRB, IACUC, IBC) outcomes (e.g. publications), contracts (e.g. collaboration, material transfer), team members and their contributions, and many more. We are excited about the future possibilities for the database, to enable more features and increase the ability to analyze the vast amount of data that accumulates on research projects. We have received much positive feedback and ideas from end-users, and we are constantly improving and expanding the system's functionality. With its adoption by all our researchers and clinician scientists, this customized and integrated database has become the critical central resource for all research being conducted, and an enabling tool for Precision Medicine.



Core **Services**

Biostatistics Core

Dr. Nasser Elkum
Director,
Biostatistics Core

The Biostatistics core serves as a hub for biomedical research in Qatar. Its aim is to advance the understanding of epidemiology and mechanisms of diseases, develop preventive, diagnostic, and therapeutic tools, and improve the health outcomes of the women and children in Qatar. It provides support across a wide range of statistical methods, data management and research designs. The core provides services to support the development and implementation of basic research and clinical trials of all sizes, collaborative clinical and patient-oriented research, and multi-institutional health services research projects.

Clinical Trials Office

Rim Imam
Manager,
Clinical Trials Office

The Clinical Trials Office (CTO) serves as the initial point of contact and centralized coordinating office for clinical research and clinical trials aiming to support the vision of excellence at Sidra Medicine. The aim of the CTO is to provide researchers and clinicians with quality services, to facilitate the design and conduct of ethical and scientifically valid research and trials and to expedite studies from initial concept to completion.

Immunogenetics Landscape of Qatari Population: From Personalized to Precision Medicine

Dr. Zohreh Tatari Calderone
Staff Scientist
Dermatology Department

With the Qatar Genome Programme, we embarked on an ambitious project to fulfill some of the objectives of Qatar's biomedical priority themes focusing on the HLA genes, containing some of the most polymorphic genes with important medical impact.

Human-Leukocyte-Antigens (HLA) genes, the main component of immunogenetics, determine the incidence and severity of viral or bacterial infections and regulate the immune response. They are also associated with several autoimmune diseases, such as diabetes. Consequently, they are used as predictive biomarkers of diseases. More importantly, the success of transplantations, both solid organ and hematopoietic stem cell transplantation, as well as gene-therapy, depends on HLA compatibility between donor/recipient.

Increasing evidence indicates the merge of immunogenetics and pharmacogenetics, where HLA-genotyping becomes a requirement to avoid adverse HLA-dependant drug reaction. The emblematic example is Abacavir, whose administration requires mandatory HLA genotyping. Many examples indicate that an increasing number of therapies will soon be tailored to a patient's immunogenetics profile for optimal drug efficacy, pushing HLA to the forefront of next-generation precision medicine.

This project would not have been possible without the effective collaboration between The Qatar Genome Programme, the Qatar Biobank, Sidra Medicine's core facilities, scientists, and clinicians. Our results indicate that HLA genotypes can capture the structure and diversity of Qatari sub-populations and provide important information leading to better allocation of public health resources and provide clinical teams with knowledge-based data to optimize the selection and distribution of biological resources to maximize patient benefit.

Research in the News

Research in the **News**

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ScaleMag – January 8th, 2020
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QU's Empower Generations Consortium signs MoUs for healthcare and education.
The Peninsula – January 24th, 2020
https://www.thepeninsulaqatar.com/article/24/01/2020/QU%E2%80%99s-Empower-Generations-Consortium-signs-MoUs-for-health-care-and-education

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Ministry of Transport and Communications – February 2nd, 2020
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Gulf Time – February 3rd, 2020
https://www.gulf-times.com/story/654954/Sidra-working-on-national-report-on-breast-cancer

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TrialSite News – February 17th, 2020
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Qatar Tribune – May 2nd, 2020
https://www.qatar-tribune.com/Latest-News/ArtMID/423/ArticleID/29383/COVID-19-Sidra-Medicine-develops-device-that-can-examine-4000-samples-a-day

Sidra Medicine successfully establishes treatment protocol for diabetes.
What's Going On Qatar – May 11th, 2020
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QF Scientists mull if immunity passport is the future option.
Qatar Tribune – November 18th, 2020
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Qatar Tribune – December 10th, 2020
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Research in the **News**

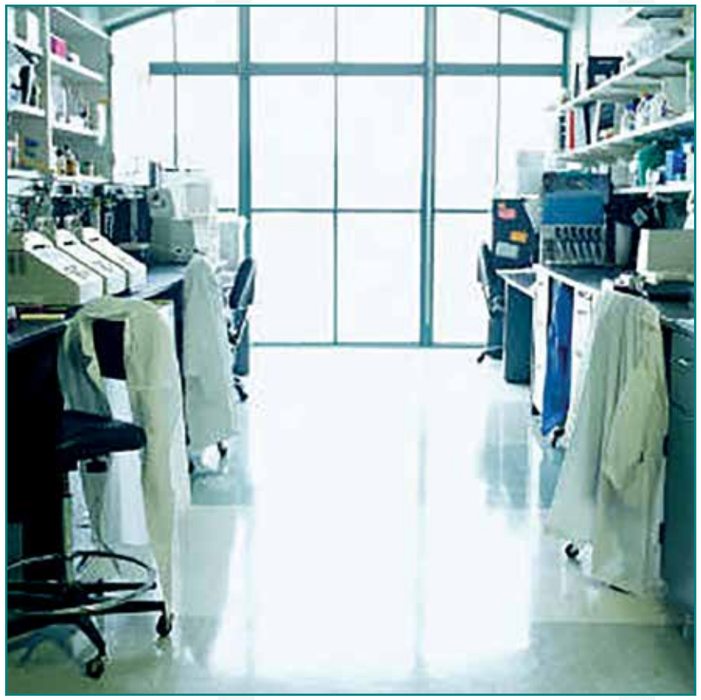
nature middle east

A silver lining for research at a time of pandemic

6th April, 2020

Scientists are using time away from their labs to dig deeply into their data.

Sedeer el-Showk



As the COVID-19 pandemic swept through Europe, governments in the Middle East quickly responded to prevent the disease from spreading in their countries. As part of this effort, many placed restrictions on non-essential businesses, services and travel. For universities, this meant switching teaching to tele-learning platforms and limiting laboratory access. Scientists, in turn, have found ways to adapt and keep research going, at least for the time being.
In Qatar, investigators at Sidra Medicine, a healthcare and research facility, are following national guidelines by working from home where possible and limiting non-essential lab work. "Most teams come in to run critical experiments on a rotational schedule to comply with social distancing principles," says Khalid Fakhro, Sidra's acting chief of research and director of precision medicine.

Nevertheless, Fakhro says Qatar's early investment in biomedical sciences is now paying off, with scientists and clinicians teaming up across institutions to tackle the pandemic on different levels, such as studying virus-host genomic interactions, creating patient bio-repositories, setting up clinical trials, and connecting with global consortia.

He adds that shifting patient visits to telemedicine has reduced the number of patients coming in for existing studies, which may delay projects planned for this year. However, his team has plenty of data to analyse from patients enrolled in studies over the past year. Without new data coming in, researchers have to work on what they already have. "
I wouldn't be surprised if this actually turns out to be a very productive year in terms of publication activity," says Fakhro.

Fahad Al Senafi, a physical oceanographer at Kuwait University, says he is also using the time to analyse data. He

says there is some benefit in researchers being forced to delve into their data rather than starting new projects, despite the slowdown in research output. “It gives people the opportunity to squeeze their data a bit more and look at things from a different perspective. You have to get a bit more creative and think outside the box, but a lot of good science could come out of this.”

Other researchers at the university, such as synthetic chemist, Talal Alazemi, have found their lab work abruptly brought to a halt and are using this time to write papers.

In Saudi Arabia, bioscientist Mo Li, who leads the Stem Cell and Regeneration Laboratory at King Abdullah University of Science and Technology (KAUST), brought some of his projects to a premature halt because of the new regulations. But he says they were fortunate to have collected large datasets before their lab went into hibernation, and will keep busy analysing them for the time being. “It will become more challenging to maintain our output if restrictions continue for an extended period,” he says.

In the UAE, New York University Abu Dhabi biophysicist, Mazin Magzoub, has come up with a time-sharing approach to ensure that work can continue. “The idea is to make sure we don›t have any more than two people in the lab at once, and it’s only really for people whose work is absolutely critical,” he explains.

His team is also taking advantage of this time to analyse and write up results and to do background reading for research.

Magzoub is also coordinating with his team to postpone experimental work while bringing forward computational work. “I’m not going to say there’s no delay. We’re not working at our normal speed, which is an unfortunate consequence but is unavoidable,” he says. “We’re compromising and trying to make the best of the situation. Ensuring everyone’s safety is the paramount concern.”

Carlos Duarte, a professor of marine science at KAUST, expects the restrictions to have very little impact on his team’s productivity unless they are extended for longer than several months. “We have a lot of work in bioinformatics, modelling, and computational biology that’s all continuing at full speed because it’s not dependent on laboratory operations,” he says. Master’s students in his group are being given alternative research projects that they can carry out under the current circumstances, and the team’s work plans will be revised every few weeks to keep pace with the situation.

Duarte’s team is also joining the global effort to learn more about COVID-19 and the virus behind it, using their genomics platform to analyse beta coronaviruses, and their spatial modelling expertise to help understand the epidemiology. “We’re using our skills to respond to this crisis, and I think everybody across campus is also contributing,” he says. “KAUST is an international university with people from 112 countries. We all have family and friends elsewhere in the world, and we want to make a difference in this global struggle.”

But Duarte has concerns. “Global attention is focused on just one thing now,” he says, meaning that a lot of valuable research “is going to go under the radar. A lot of science that would have influenced policymaking and decision-making is going to be missed.”

Fakhro says scientists in Qatar also see working on COVID-19 as a priority. At Sidra, expertise in single-cell RNA sequencing and high-throughput genomics have been combined to develop an innovative testing strategy for the virus. “As an academic medical centre, Sidra’s genomics research core is working closely with counterparts in clinical pathology to develop testing solutions that are as sensitive as clinically approved standard tests, while offering increased throughput and faster results,” he says.

“While the current crisis imposes the need to stay safe and maintain distance, never in the history of humanity have we felt closer to each other than this time, as we come together as a global scientific community to face this challenge head-on,” says Fakhro.

Research in the **News**

The Peninsula
Local focus, Global vision
QATAR'S DAILY NEWSPAPER

Sidra Medicine uses precision medicine to treat children with rare diseases

10th November, 2020

Dr. Fakhro (left) Dr. Amel Hassan (right)

Doha: Sidra Medicine, a member of Qatar Foundation, aims to bring to the healthcare landscape in Qatar through application of precision genomic medicine especially in the case of rare diseases in children.

When a child starts showing symptoms that are unheard of or they don’t fit the description for a commonly known disease, a phenomenon known as the ‘diagnostic odyssey’ follows, where the patient will on average spend
about five years, visit at least seven
different physicians, undergo a myriad of tests, and in many cases, travel the world seeking answers to their symptoms.

The delay in diagnosis not only costs the patient in terms of health outcomes but is also very costly for a medical system to bear. Instead, the ability to receive an accurate diagnosis in a short time frame could dramatically improve quality of life for these patients and provide opportunities for treatment at an early stage.

Sidra Medicine currently performs genome sequencing for patients across a wide range of research studies. Genome sequencing is the practice of decoding a genome – which is an organism›s complete set of DNA including all of its genes. It is a valuable diagnostic tool that allows researchers to determine if any disease-causing mutations are present.

“Sidra Medicine is an academic medical center where education, research and clinical practice converge to deliver the best care for women and children. It’s where scientists and doctors work alongside each other, supported by cutting-edge technologies, to help find answers, and give our patients hope and confidence where it may not have been seen before,” said Dr. Khalid Fakhro, Acting Chief Research Officer at Sidra Medicine and Director of its Precision Medicine Program.

In July 2019, a baby suffering from severe infections and poor muscle tone since birth visited Sidra Medicine. Frequent infections had resulted in injuries to her gut and lungs, due to which she repeatedly required assisted ventilation. The illness had rapidly taken a toll on her young body, resulting in loss of weight and a lack of growth. The child underwent clinical genetic testing by sending their samples abroad to a commercial lab; however, the mutations identified were not particularly known to

be disease-causing. The patient was then referred to research.

In a multidisciplinary effort, Dr Amel Hassan, Senior Attending Physician in Allergy and Immunology, and Dr. Bernice Lo, Principal Investigator in Human Genetics, both at Sidra Medicine studied the patient's white blood cells. They found that the patients' immune function was impaired and that a variant in the gene ORAI1 could explain the child's condition.

Further investigation, in collaboration with Dr Khaled Machaca's lab at Weill Cornell Medicine in Qatar, confirmed that the mutation present in ORAI1 causes a functional defect. Fortunately, there is already treatment available for ORAI1-related immune deficiency. Within days, the child was put on a new treatment plan, which significantly improved her clinical condition until she was healthy enough to leave the hospital for the first time in seven months.

Dr. Hassan said: "If left untreated, this condition would have resulted in the passing away of the child before her second birthday. There are different options of potentially curative treatments but defining the exact molecular or genetic defects is crucial in tailoring the appropriate treatment, maximizing chances of survival, and improving quality of life.

"Working closely with a dedicated research team enabled us to confirm the diagnosis and explain to the parents the reason behind their child's clinical symptoms. Having successfully diagnosed her condition, we were then able to administer a targeted treatment, which allowed her to finally leave the hospital, after having been an inpatient for seven months.

"The ability to perform genetic testing at Sidra Medicine allowed us to diagnose the child's condition in a timely manner thereby preventing the risk of multi-organ complications, which could have proved to be fatal. A similar diagnosis, five years ago, would have necessitated a trip abroad."

Rare diseases are predominantly caused by genetic mutations. Over 50 percent of rare diseases affect children, 30 percent of whom die before the age of five due to a lack of diagnosis and treatment options. Genomic medicine is key to unlocking the cause of rare diseases, and to prescribing appropriate treatment in the future.

Through its efforts, Sidra Medicine remains committed to taking research from bench to bedside. They do this by eliminating the barriers between research and clinical practice and allowing their researchers and physicians to work side-by-side.

"At Sidra Medicine, we have established a center of excellence in genomic medicine. In the coming years, we aim to make genome sequencing available to all our patients. Imagine every patient having their genome sequenced and already in their medical record. We can then predict disease risk, know a patient's drug sensitivities, and tailor treatment to an individual's genome. That would be transformational; that is true Precision Medicine," Dr. Fakhro said.

Research in the **News**

مؤسسة قطر
Qatar Foundation

Using fish to diagnose diseases in humans

2nd December, 2020

Researchers at Sidra Medicine, a Qatar Foundation medical and research center, are working with zebrafish to understand human disease, particularly in children

A little-known fact is that several species of fish have genes that are extremely similar to those of humans. One such fish is the zebrafish. Small and unassuming, the zebrafish shares 70 percent of its genes with humans making it an ideal organism to help further our understanding of human genetics and diseases.

This is exactly what the Zebrafish Functional Genomics Facility at Qatar Foundation's Sidra Medicine is currently working on. Using these small organisms, researchers are able to make connections between genetic variations and the diseases they cause

When patients approach Sidra Medicine with unusual clinical presentation that leads to inconclusive diagnoses, they are referred for genetic testing. Although specific tests are conducted in the different clinics depending on the nature of their symptoms and suspected conditions; unsolved cases get referral for genetic testing.

The results of the genetic testing help identify some novel variants that have not been reported before. Knowing that a novel variant exists is not enough information to determine that it is the cause of a patient's symptoms; so this data is provided to the zebrafish facility for research. The facility uses it to recreate a mini fish model of the patients' genetic variation to study its relation to the disease..

Being able to recreate it in the zebrafish model, give us a better understanding of whether or not this really is the case. This allows for families to have a better understanding of what is happening

> **Dr. Sahar Da'as**

The facility currently conducts research primarily on rare pediatric conditions of congenital abnormalities, neurological disorders and cardiovascular diseases.

"When we find genetic mutation or variation, we cannot approach a patient's family and tell them that this is the cause of their child's disease, as we cannot be sure. Being able to recreate it in the zebrafish model, give us a better understanding of whether or not this really is the case. This allows for families to have a better understanding of what is happening, as well as for clinicians to decide which treatments or methods of management would be most effective," said Dr. Sahar Da'as, Laboratory Manager at the Sidra Zebrafish Facility. What makes zebrafish ideal for research like this – apart from easy accessibility and their small size – is that a large percentage of

their genes also simulate human disease.

A specific case that stands out to Dr. Da'as is one where a child arrived at Sidra Medicine with a rare, complex neurological disorder that only has 50 reported cases worldwide. Upon detailing family history, doctors at Sidra Medicine found that the child's mother had been complaining of reduced fetal movement while she was pregnant. This complaint had not been identified in any of the other cases. This allowed researchers to recreate the genetic variant found in the child in zebrafish model and look for this unique symptom.

Our goal at the moment is to understand the genetic mutations that present most commonly among the pediatric population

Dr. Sahar Da'as

Sure enough, they found that when replicated in zebrafish, the variant caused brain abnormality, reduced motor skills and muscle movement at very early stages of development. While in this case, unfortunately, this did not offer much in the way of conclusive therapies, it did help clinicians understand the cause in order to advise the parents on how to manage future pregnancies and seek help early.

Sidra Medicine also uses zebrafish for research in pediatric diabetes, cancer, and cardiac disease. According to Dr. Da'as, the fish lend themselves well to research for a number of conditions. "Our goal at the moment is to understand the genetic mutations that present most commonly among the pediatric population. While we aim to conduct research on conditions and mutations that present in pregnant women and babies in-utero, we hope to also work on infertility issues in the future," she said.

Distinct Antibody Responses Against "Common Cold" Coronaviruses in Children and Adults

Dr. Nico Marr
Principal Investigator
Immunology Department

Four endemic human coronaviruses are commonly associated with acute respiratory infection in humans. Antibody responses to these "common cold" coronaviruses remain incompletely understood. Harnessing the unique resources of Qatar Biobank and in collaboration with members from Sidra Medicine's Pathology Department, Qatar University and international collaborators, we have performed a comprehensive analysis of coronavirus-specific antibody responses in 231 children and 1168 adults using a novel technique called phage-immunoprecipitation sequencing. Interestingly, we found antibody responses against these viruses to be qualitatively different between children and adults in that those antibodies more frequently found among children targeted functionally important and structurally conserved regions of the viral proteins. Moreover, some antibodies that bound to a particular region in the spike protein of endemic HCoVs were broadly cross-reactive with proteins of other coronaviruses, including SARS- and MERS coronavirus. Our findings shed light on the targets of human antibodies that are specific to certain coronavirus species and those that are shared between different coronaviruses, thereby providing important insights for the development of antibody therapies and vaccine design. Such antibodies may affect the dynamics of sporadic MERS outbreaks that mostly occur in the Middle East, and the current COVID-19 pandemic. Building on the Qatar Genome Programme, we are currently also exploring how human genetic factors can shape antibody responses in health and disease.

Importance of functionally validating variants of uncertain significance

Dr. Amel Hassan, Dr. Bernice Lo and their team Dr. Rafah Mackeh and Dr. Nourhen Mansour

"Studying the functional consequences of variants is important to determine if the variants are disease-causative and provide an accurate diagnosis. An early accurate diagnosis enable the patient to be provided the proper treatment and care improve health and quality of life."

Dr. Amel Hassan, Senior Attending Physician in the Allergy and Immunology Department at Sidra Medicine, specializes in treating patients with inherited immune deficiency disorders. One baby presented with frequent severe infections, poor muscle tone and strength, and inability to sweat. The frequent infections were causing worsening of health with loss of weight, lack of growth, and injury to the lungs and gut. The clinical genetic testing for the patient only identified variants that were unknown whether they were disease-causative. Dr. Hassan astutely determined that the variant in the gene ORAI1 could potentially explain the patient's clinical picture. In collaboration with Dr. Bernice Lo from Sidra Medicine and Dr. Khaled Machaca from Weill-Cornell Medicine-Qatar, the functional consequence of the variant was assessed. The mutant ORAI1 was found to be defective and did not even localize to the correct position in the cell. This defect resulted in an impaired immune response by the patient's white blood cells. These data helped confirm that the variant in ORAI1 is indeed pathogenic. Dr. Hassan therefore provided the treatment appropriate for ORAI1 immune deficiency, which significantly improved the child's clinical condition so that the child was healthy enough to leave the hospital and go home for the first time in 6 months.

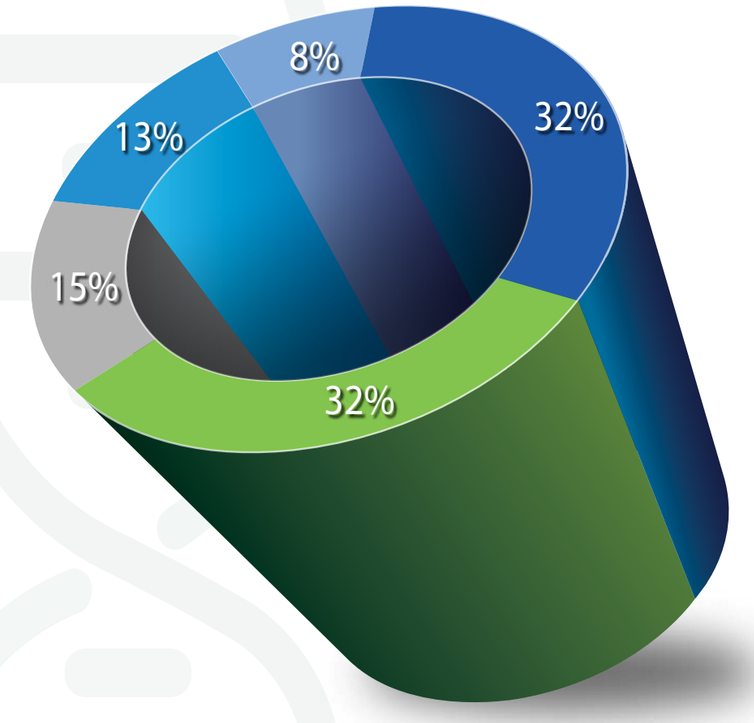
SIDRA MEDICINE RESEARCH ANNUAL REPORT 2020

Collaborations and Grants

Collaborations

Sidra Medicine Research collaborates with institutions both locally and internationally. Sidra Medicine Research's commitment for collaborating internationally with prestigious organisations is equal to the effort put in enriching local institutions inside Qatar. About half of all research projects benefit from such collaborations.

International Collaborations

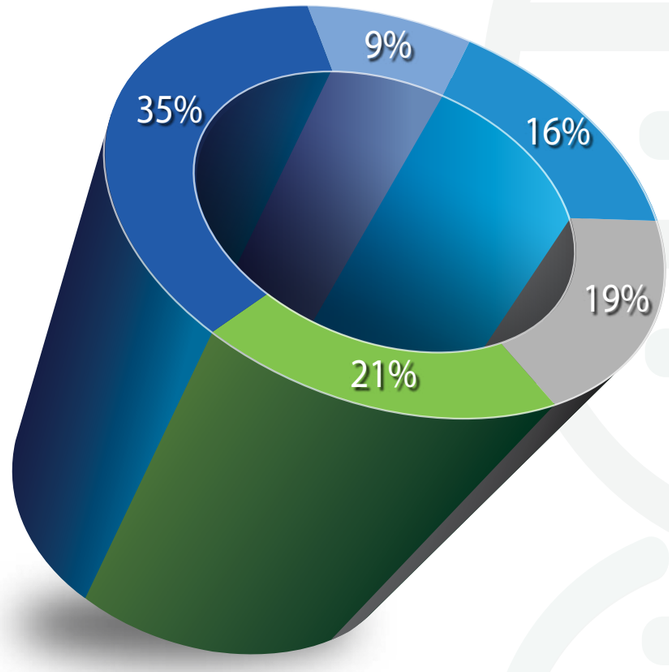


United States
United Kingdom
Spain
Italy
Other

Australia
Turkey
Germany
Greece
Pakistan
France
Canada
Brazil
Hungary
Lebanon

Sidra Medicine Research's local collaborations fosters Qatar National Vision in building a knowledge-based economy in biomedical and health sciences. This ensures knowledge transfer from institutions considered thought leaders in these areas and builds sustainability in biomedical research. Sidra Medicine is invested in building productive partnerships with all local biomedical institutions. Across our portfolio of projects we have several local collaborations. Together we strive to create sustainability for biomedical research in Qatar.

Local Collaborations



Hamad Medical Corporation
Hamad Bin Khalifa University
Weill Cornel Medicine-Qatar
Qatar University
Other

Ministry of Public Health
Texas A&M University -Qatar (TAMU-Q)
Qatar Museums Authority
Anti-Doping Lab Qatar (ADLQ)
Equine Vetrinary Medical Centre (EVMC)
Primary Health Care Center (PHCC)

Grants

From the numerous research collaborations, Sidra Medicine Research works closely with Qatar National Research Fund (QNRF). In 2020 alone, Sidra Medicine has been awarded 18 projects, out of which 4 are clinically driven. The projects amount to QAR 17,268,194. Projects awarded to Sidra Medicine include:

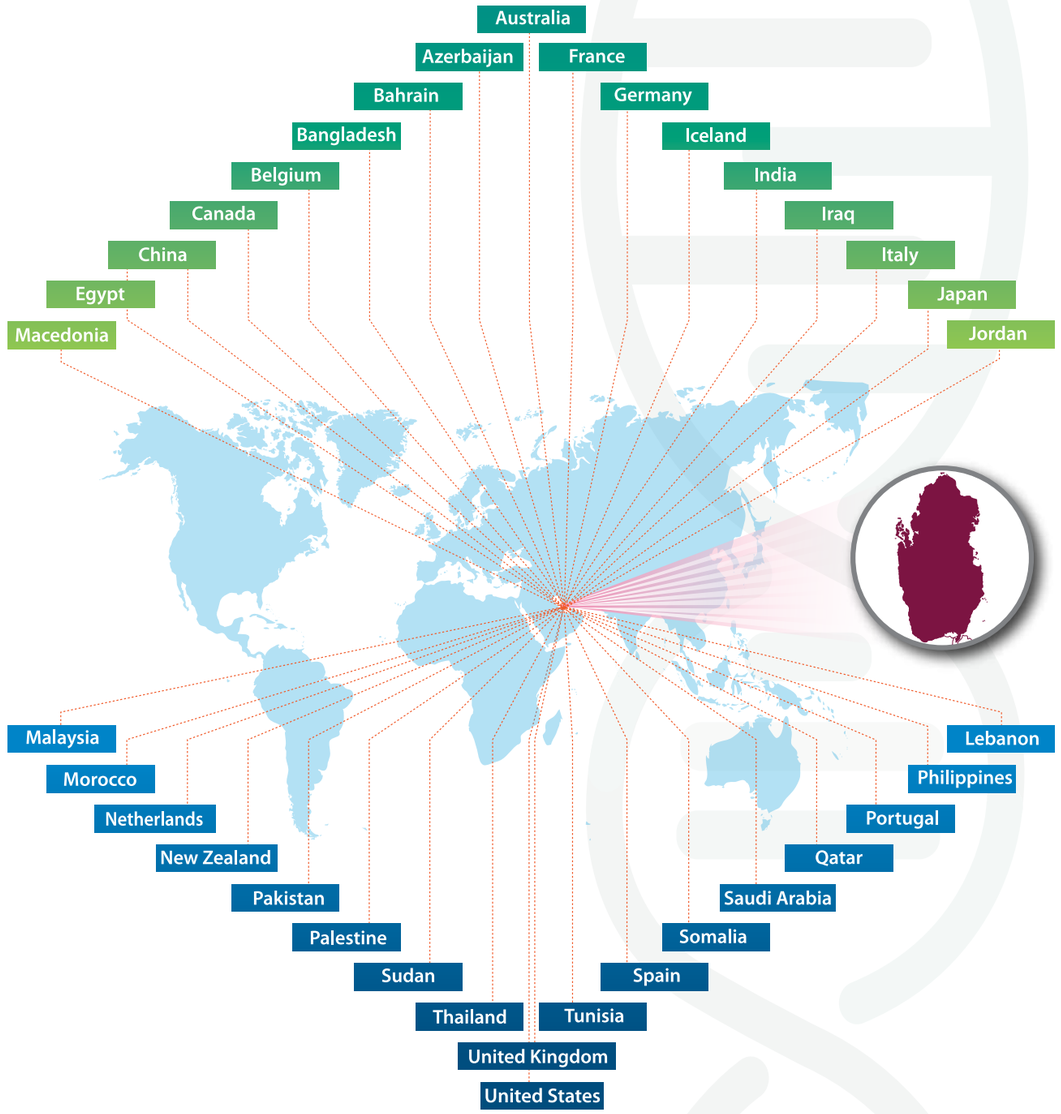
- National Priorities Research Program (NPRP)
- Path Towards Precision Medicine (PPM)
- Conference and Workshop Sponsorship Program (CWSP)
- Researchers Exchange and Mobility Program (REMP)
- Junior Scientist Research Experience Program (JSREP)
- Early Career Researcher Award (ECRA)
- Rapid Response Call (RRC)

Through QNRF, Sidra Medicine also co-funds projects both locally and internationally.

Grants	Awarded Amount	
NPRP	QAR	2,168,582
NPRP	QAR	2,166,870
NPRP	QAR	1,994,476
NPRP	QAR	1,861,500
NPRP	QAR	2,080,062
NPRP	QAR	2,183,269
PPM	QAR	1,639,543
PPM	QAR	1,751,299
CWSP	QAR	127,750
CWSP	QAR	127,750
CWSP	QAR	10,950
REMP	QAR	444,500
REMP	QAR	34,875
REMP	QAR	101,265
REMP	QAR	27,375
RRC	QAR	61,320
Amryt (external grant)	QAR	245,000
ECRA	QAR	492, 808

Nationalities at **Sidra Medicine Research**

35 Different nationalities currently working at Sidra Medicine Research



The Baraka Study for Autism

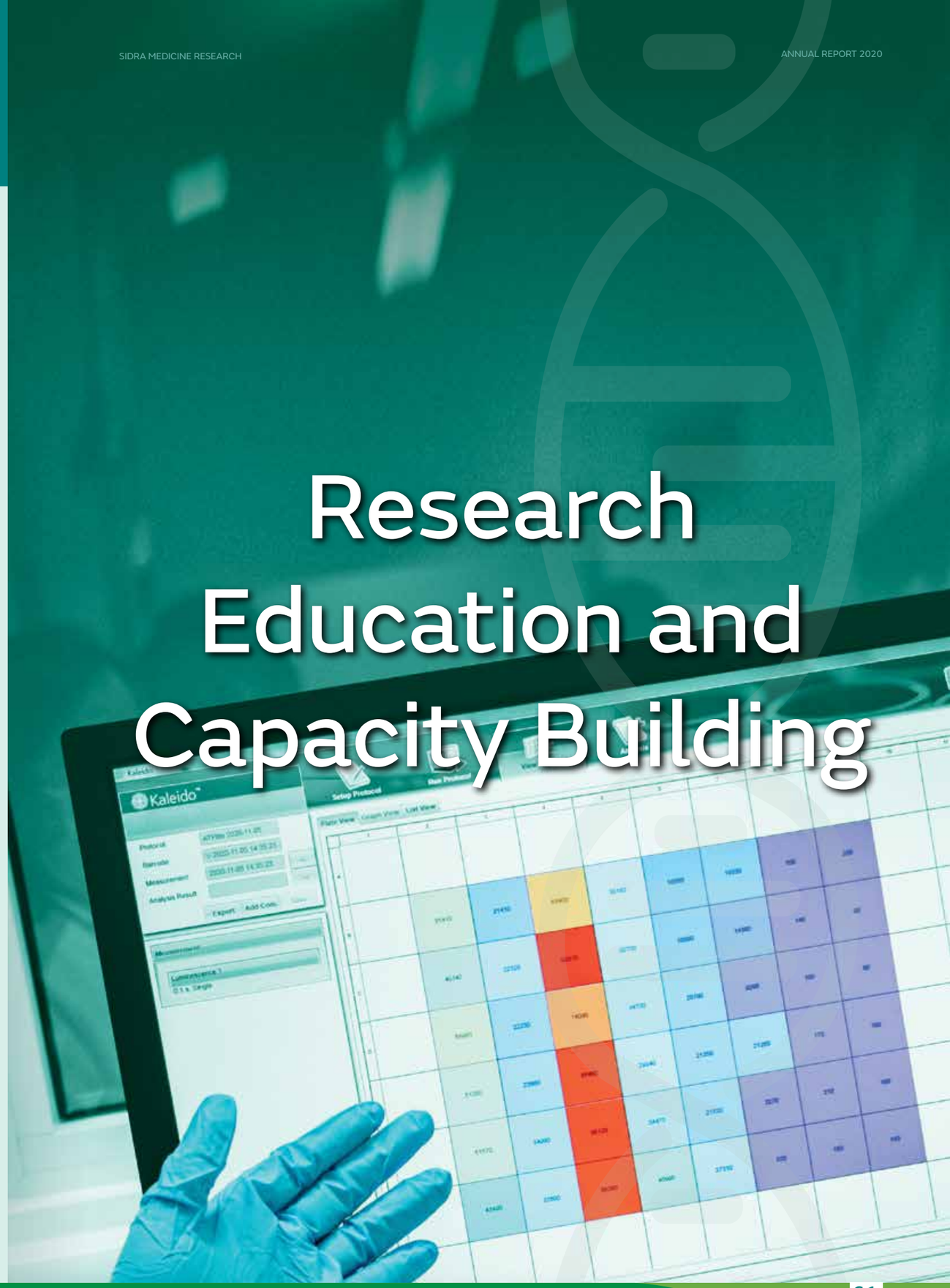
Dr. Madeeha Kamal
Senior Attending Physician
Adolescent Medicine

"Our kids with Autism are going to be adults with Autism. We need to help them overcome obstacles on the spectrum. This study is a great help for kids not only in the country, but in the region and across the world"

Autism Spectrum Disorder, once thought to be a rare disease, is now incidentally a remarkably common disease, estimated to affect at least 1 in 68 children. ASD is a heterogeneous neurodevelopmental condition comprising of social, behavioural, communication and in some cases, cognitive abnormalities.

Dr. Madeeha Kamal, a Senior Attending Physician in the Adolescent Medicine Department at Sidra Medicine, is one of the principle investigators in the Baraka Study, a study which deeply explores Autism Spectrum Disorder (ASD). The name Baraka is the Arabic word for blessing, carrying the dual cultural connotations, of attaining divine blessings in the quest to study ASD in Qatar and that all children regardless of developmental challenges are a blessing. The study is QNRF-funded and is in collaboration with SickKids in Toronto, Canada. The main goal of the study is to create a long-term National Resource for ASD research, which will help usher in an era of scientific pursuit of both environmental and molecular risk factors for ASD in Qatar. To achieve this, the study tackles the disorder on three fronts; Clinical, Genomic and Translational. The study uses patient samples more deeply using multiple omics technologies. Mutations or pathways discovered from patients are then followed-up by using model systems to better understand the pathophysiology of the disease. The study aims to enrol 2,000 patients and their familes between Sidra Medicine and Hanad Medical Corporation to characterize them and develop personalized therapy for them in the future. Incorporating elements of precision medicine is crucial for the Baraka study. It helps answer the unreciprocated questions that clinicians face whilst observing response to medication to each individual ASD patient.

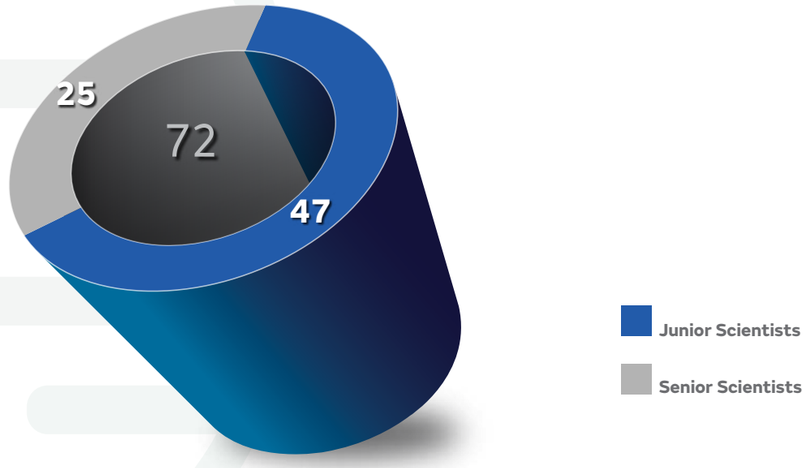
Research Education and Capacity Building



Research Education and Capacity Building

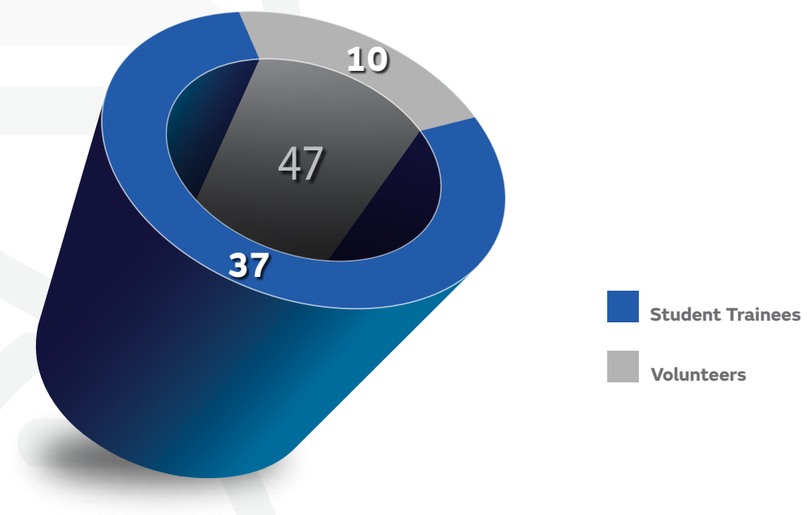
Outreach and education at Sidra Research is paramount in providing support to Post-Doctoral Fellows, Research Scientists, Graduate Associates and Trainees. Educational talks are an integral part helping them identify their interests and to further develop their skills. Sidra Medicine's flagship Research Series, reinforces support, growth and development amongst researchers and is a first-hand source of education for our future scientists who are training at Sidra Research.

2020 Educational Talks



The research department hosts several trainees, volunteers, and visiting researchers/scientists coming from various universities and institutions, both locally and from around the world. Sidra Research prides itself as a teaching entity in alignment with the education pillar, one of the three pillars that form the foundation of Sidra Medicine's mission to provide patient care and biomedical research. Training at Sidra encourages young adults to hone their career path in science, medicine and public health. By training under varied professionals and experts, the trainee is provided with invaluable work experience, develops and refines skills and has access to a platform to network with other professionals in the field.

Trainees and Volunteers



Research Education and Capacity Building

Moza Al Kuwari-HBKU Alumna Underlines the Importance of Medical Research

Moza Al Kuwari
Research Specialist

PhD student's educational journey prepared her for medical challenges of today in a story covered by QF.

Pursuing higher education had always been Moza Khalifa Al Kuwari's dream. And her long-spanning educational journey – which saw her start her bachelor's degree at Qatar University and recently conclude her PhD at Qatar Foundation – has helped prepare her to face the medical challenges of today.

Driving Al Kuwari's educational journey is the belief that research is imperative to creating solutions to worldwide medical issues, such as COVID-19. "I believe that the importance of research has become very evident, especially in light of the current crisis, which has forced us to rethink the way we look at things," explains Al Kuwari.

In between periods of studying, Al Kuwari worked at Hamad Medical Corporation, as well as the Qatar Biomedical Research Institute at Hamad Bin Khalifa University (HBKU), a member of Qatar Foundation (QF). Now, she is a research specialist at Sidra Medicine – also a member of QF – studying the molecular basis of hearing loss.

Al Kuwari highlighted the role QF has played in her career, saying: "I already have great work experience and diagnostic skills that I acquired from working at leading medical institutions. But at Qatar Foundation I was also able to expand my knowledge of medical research, which is completely different to the diagnostic field, and has enriched my experience in research labs.

"As being part of the QF family, I was given countless opportunities to participate in research events, such as the Qatar National Research Fund's Annual Forum, and other conferences organized by QF entities. This, in turn, gave me the chance to meet international experts, opening doors for collaboration in the field of research."

Al Kuwari completed her BSc in Biomedical Sciences at Qatar University in 2004, and, while dedicated to her further studies, was reluctant to travel abroad to pursue her postgraduate studies, as she did not want to be away from home. "However, years after my graduation, Qatar University launched a new Master's program Biomedical Sciences, and I was one of the first students to enroll, so was among the first batch of graduates," Al Kuwari said. Similarly, when HBKU's College of Health and Life Sciences announced the launch of the PhD program in Biological and Biomedical Science, Al Kuwari did not think twice about applying.

Describing her PhD experience, Al Kuwari says that it was very challenging, not only in terms of the academic side or balancing responsibilities between studies, work, and family, but also because the program was new.

"When I joined in 2015, it was still new, and we had to work together to create the foundations of the program, as well as the research labs and facilities, and pave the way for the next batch of students. I feel very proud to be among the students graduating from this program," Al Kuwari explains.

Today, Al Kuwari is focused on the functional and molecular characterization of hearing loss in Qatar by tackling the hearing loss gene through data collected by the Qatar Genome Programme and patients attending the ENT clinics at Sidra Medicine.

In the future, Al Kuwari is looking forward to continuing to work in the field of biomedical sciences through identifying the genetic causes of disease and understand its pathophysiology. And, importantly, to transferring her knowledge and working experience to the next generation of medical students.

Republished from:
https://www.qf.org.qa/stories/hbku-alumna-underlines-the-importance-of-medical-research

Research Education and Capacity Building

Sheema Hashem – On Working as a Research Specialist

Sheema Hashem
Research Specialist

My name is Sheema Hashem, and I work as a Research Specialist at Sidra Medicine. I received my B.S. in Chemistry from Qatar University, but at Sidra Medicine, I started working in the field of cell biology, where I combine the chemical and biological approach towards developing some novel technology that could benefit humanity in the long run.

All the scientific advancements, whether small or big, make me extremely excited and encourage me to dig more into scientific research to discover some novel treatments for debilitating diseases, and one of them is diabetes. In the future, I would like to work towards contributing to significant scientific advancements that can cure diabetes. My passion for this comes from the pain of seeing people who are close to my heart suffering from this disease. I want to see some significant scientific advancement which can cure diabetes entirely and should be less painful and affordable to the patients. Science is striving to find a treatment for diabetes but how close are we? Biotech and pharma industries are striving hard to develop new type 1 diabetes treatments. A few achievements include;

1) Swapping missing cells with cell therapy: Although still in the very early stages of development, cell therapy is one of the biggest hopes towards developing a cure for diabetes, especially for type 1 diabetes.

2) Immunotherapy as a means of a cure: There are organizations that are working towards developing a vaccine for type 1 diabetes that stimulates the immune system to lower the levels of an inflammatory protein that is thought to be involved in multiple autoimmune diseases.

3) Artificial pancreas as an automated means of treatment: For people that have already lost their insulin-producing cells, a shorter-term solution could be the 'artificial pancreas' — a fully automated system that can measure glucose levels and inject the right amount of insulin into the bloodstream, just like a healthy pancreas would.

The approaches to cure type 2 diabetes that is either currently going or under process include;

1) Stimulating insulin production: Here again, the companies use different strategies to stimulate insulin production for the cure of type 2 diabetes. One of the biggest hits is glucagon-like peptide (GLP)-1 receptor agonists, which induce insulin production in beta-pancreatic cells while suppressing the secretion of glucagon.

2) Gut microbiome as a target to cure type 2 diabetes: Although promising, the microbiome field is very young, and until more diabetes treatments are tested in the clinic, it will be challenging to determine the real potential of the microbiome in this space.

While working at Sidra Medicine I learn more and more about this matter every day and my hope that one day a great scientific breakthrough will be made for diabetic patients grows. I hope in my lifetime to see a form of treatment for diabetes, where the body can produce its own insulin.

I want to see a scientific advancement where we can change the flexibility of the cells to adapt them to different identity and perform the function of damaged cells. If we gain more knowledge about the mechanisms behind this cell flexibility, then we could possibly be able to control the process and change more cells' identities so that more insulin can be produced. This achievement would save and change the lives of countless amounts of people, and this is what keeps me going and passionate about my work.

Research Education and Capacity Building

Alia Al Massih - Graduate Associate

"As a Qatari graduate at Sidra Medicine, I have the desire to enrich my understanding and knowledge by pursuing further studies and tracking scientific developments. I believe through this, I will be able to benefit not only society but also public health"

Alia Al-Massih
Graduate Associate

Sidra Medicine offers prospective Qatari scientists an opportunity to benefit from the broad range of institutional expertise and begins preparing them for the challenges of an ever-changing world. Sidra Medicines sponsors Qatari students to complete their studied, and return back to Sidra for a year of exploratory experience, where they are exposed to several different facets of research, before welcoming them as a full time scientist.

After being enrolled in the National Graduate Development Program, Alia had the chance to improve her skills and knowledge and attended various online science conferences and courses.

"Within one year as a graduate associate (GA) in the Research Department, I attended seminars, workshops, and two Sidra Medicine conferences: Current Understanding in Diabetes, Obesity, and Related Syndromes (CUDOS); and The Maternal and Child Health Symposium (MCH). Besides that, I made three lab rotations with different Principal Investigators and got involved and exposed to researchers' projects, and learned other lab techniques, kits, and machines.

Additionally, I had one rotation with one of the Biomedical Informatics teams and attended training sessions. Moreover, being treated as a permanent team member, not a trainee or graduate associate, meant I could attend team meetings, journal clubs and present my work and lab results to the team members for discussion and comparison." After achieving a top 5 ranking among Qatari graduates in 2015, Alia Al Massih began achieving her dreams by accepting an offer to attend the University of Leeds, a member of the prestigious Russel Group association of research universities. Alia noted, "Those steps were the lead for extending my aims to apply for Sidra's Medicine scholarship program, which *Alhamdulillah* I achieved after passing the interview and successfully earning the required criteria. Attaining such an opportunity from a well-respected institute with high standards and a well-studied strategic approach would play a significant role in helping me propel towards greater achievements.

Studying abroad and earning a degree in Medical Biochemistry from a creditable institute within the Russel Group was a big challenge."

Testimonials from **Trainees**

Nutrigenetics Externship

During my first year of graduate studies at Hamad Bin Khalifa University, I met several adjunct and joint professors who were researchers at Sidra Medicine. Through them, I learnt about the various research projects, advanced technologies and facilities that are available at Sidra Medicine. I was particularly interested in Dr. Annalisa Terranegra's research in the nutrigenomics field since I come from a nutrition and dietetics background. This was how I started learning more about the training opportunities that Sidra Medicine offers. This led me to join Sidra Medicine for my thesis' research in June of 2019 as a research trainee for a one-year program. My experience was useful, productive and memorable. I had no previous experience in a gut-microbiome lab, so my mentors Dr. Arun Lakshmanan and Dr. Dhinoth Bangarusamy taught me all the lab techniques and protocols that I needed for my project from scratch. My supervisor and my mentors were always available and supportive. They equipped me with the laboratory and analytical skills needed to complete my master›s thesis. Moreover, they were keen to listen and apply my ideas particularly related to the field of dietetics. I was also given the opportunity to write abstracts which were accepted in two conferences for a poster presentation. All of this contributed to building my research identity as a young researcher and to follow my passion in biomedical research. The project I worked on included type-1 diabetic and type-1 diabetic-obese paediatric patients. Although finding a causal association was not the objective of my thesis project; however, my interest in the coexistence of diabetes and obesity phenotypes continues till today. The highlight of my experience is the friendship I developed with my fellow trainees in the lab. It will be my strong advice for future trainees to get the best out of their time at Sidra at all levels; it is a great scientific hub to gain knowledge and skills through training and workshops, networking, and of course fun. I urge future trainees to be proactive learners instead of passive, to seek advice from experienced researchers, and not to be shy to express their thoughts and ideas. I recommend they strive for efficiency and take initiative. Finally, I believe that they should try to leave their imprints on their work and in the workplace.

Farah El Assadi
Hamad Bin Khalifa University
Masters Student Trainee under
Dr. Annalisa Terranegra

Testimonials from **Trainees**

Cancer Immunology Externship

My ambition and desire to be a qualified and skilled researcher has led me to pursue an externship program at Sidra Medicine, particularly the Research Department for several reasons. Sidra is a leading research institute not only locally, but worldwide as well, with highly qualified researches and staff, a supportive environment that values continuous learning, in addition to well-prepared laboratories and facilities. Given that, I strongly believe that Sidra will be the best place to achieve my goals. I joined Sidra externship program as an extern in Functional Omics Lab led by the principal investigator Dr. Wouter Hendrickx. The research group combines the best of both worlds, wet lab, and dry lab. In wet lab, I joined an ongoing project; SLFN11 Modulation in Breast Cancer Cells Affects Toxicity of DNA Damaging Agents, where I have learned new techniques and gained more knowledge. On the other hand, I was curious to learn dry lab skills. I have been frequently exposed to interesting bioinformatics projects and presentations in group lab meetings and during conferences. This has opened my mind and made me passionate about bioinformatics and motivated me to discover this field. Bioinformatics has great tools to analyze biological data generated from different experiments such as whole genome and RNA sequencing, and it is at the core of scientific discovery. I believe that data analytics skills are essential for biologists. In fact, biology is evolving into a data science. Therefore, I am considering a combined career between wet and dry lab. Getting started with bioinformatics was challenging as this field is totally new to me and combines many disciplines including computer science, statistics, and biology. However, by the guidance and support from my PI and colleagues I was able to start learning bioinformatics, in particular R-programming to perform cancer transcriptomic studies. Overall, I am proud and satisfied with what I have achieved at Sidra, and it has really been a great experience with such a great PI and team. My advice for the future externs is to make the best of this great opportunity, work hard to achieve your goals and enjoy the journey!

Eiman Ibrahim Ahmed
Qatar University
Student Trainee under
Dr. Wouter Hendrickx

Testimonials from **Trainees**

Zebrafish Core Facility Externship

My story with Sidra Zebrafish Core Facility started in 2019, when I participated in the "Zebrafish Workshop, hands-on" organized as part of the third annual "Current Understanding of Diabetes, Obesity, and Metabolic Syndromes (CUDOS)" congress that took place between 20-26 November 2019.

In the 2-hour workshop, I was intrigued by the Zebrafish model, and wanted to know more about using this model in research. I got the chance to meet with Dr. Sahar Da'as and her team and we planned for an extending training at the facility. When I applied for the externship, the administrative staff at Sidra Medicine handled my application with care and made me feel very welcomed.

After being accepted, the time I spent at the Zebrafish facility was very fruitful. The facility is well-organized and very well equipped. The brief training program was well-structured and executed. I had the opportunity to learn a plethora of new hands-on skills which includes the use of gene editing tools to manipulate zebrafish genome in order to examine the mutational effect of certain genes on zebrafish phenotype. This motivated me to plan for future in vivo research that involves animal models as part of my dissertation.

Based on what I have experienced during this training, I would highly recommend students to apply for training programs at Sidra research facilities as the experience they will get there is unique and can be compared to no other.

Dr. Sahar Da'as and her team were very pleasant, helpful, and supportive. I would like to thank them and Sidra Research for hosting me during this externship program and I am looking forward to visiting Sidra Medicine again for other training opportunities in the near future.

Wesam Shafiq Ahmed
Hamad Bin Khalifa University,
PhD Student Trainee
Under **Dr. Sahar Da'as**

Testimonials from **Trainees**

Human Genetics Externship

I was excited to join Sidra Medicine due to its advancement in the research field of Genetics and the involvement of patients in the research process, which seemed remarkably interesting and promising to me. During my time at Sidra Medicine, I was training under the Human Genetics Department, especially with the Mendalian disease cases. During which, I was exposed to various aspects of biomedical research which piqued my interest, such as data analysis. This led me to a deeper understanding of the research process and its various building blocks and inspired me to pursue working in the NGS data analysis field. Whilst working in this field, I was able to acquire knowledge and apply it, where me and the team in Dr. Khalid Fakhro's lab were able to come out with unique and informative research outcomes which will result in a publication.

I would advise future externs to make the best of their experience at Sidra, and be open to acquire as much knowledge and experience from the experts in many evolving fields that are under one roof.

Sana Al Saafin
Hamad Bin Khalifa University
Master Student
under **Dr. Khalid Fakhro**

Educational **Events**

Sidra Medicine Research Participates in CMU-Q Bio Career Day

Sidra Medicine participated in Carnegie Mellon University's Bio Career Day where several students aspired to learn more about opportunities our academic research hospital offers. The students were highly engaged and displayed an exceptional level of ambition and interest at the possibility of interning at Research Branch for experience and exposure to lab work.

Zebrafish Model for Precision Medicine

Dr. Sahar Da'as
Laboratory Supervisor for the Zebrafish Facility

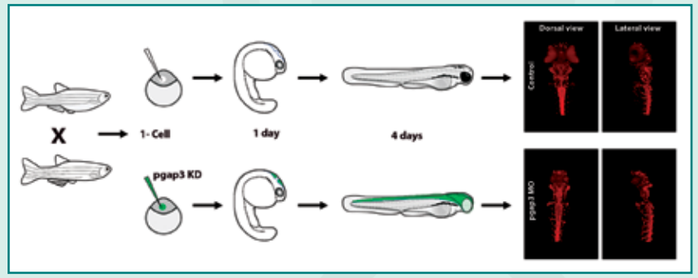
The Zebrafish Functional Genomics Core Facility is a critical hub for translational research unlocking the mysteries of human disease through precision medicine. A wide variety of diseases such as neurological disorders, developmental disorders, muscular dystrophies and heart conditions, result from mutations in human genes.

Interestingly, the Zebrafish model at Sidra Medicine helps researchers create patient-specific genetic models. Gene-editing technologies, used in this facility, offer a new level of accuracy and specificity to precisely manipulate these genes to develop a fish version of the studied human disease. The physical transparency during the zebrafish's embryonic and larval stages has been a key factor in allowing researchers to observe developing organs within a few days of studying the difference between health and disease.

Over the past year at Sidra Medicine, the zebrafish facility has aided clinicians at the Neurology clinic to discover the nature of a very rare complex neurological genetic condition that is associated with mental disorder reported in only 50 patients worldwide. The zebrafish patient-specific model showed the effect of the patient PGAP3 gene variation on brain shape and muscle movements successfully within five days. The studies explain the impact of this variation originating at very early stages of the fetal brain and central nervous system development during pregnancy, thereby affecting the nerves through the spinal cord.

Recently, zebrafish research led to solving a new genetic cause of human motor neuropathy disease. We examined the novel disease VWA1 gene shared in 17 patients. The specific fish model highlighted that the causative gene alteration resulted in complex biological rearrangements leading to degeneration of the developing motor neurons affecting the neuron connections to the muscles and thus leading to muscle weakness in the affected individuals.

"Our unique facility platform deciphers the scientific commitment for achieving an influence on the patients' wellbeing. Utilizing zebrafish models creates a greater understanding of human genetic disorders that will guide clinicians towards better patient management helping families better understand their child›s condition" said Sahar Da›as, the facility manager. Looking to the future, researchers at Sidra will continue to improve approaches to develop zebrafish-specific disease models and use these models of patient-specific mutations for developing future individualized treatments.



Zebrafish muscle fibers showing actin filaments labelled with green fluorescent protein and neuromuscular junctions labelled with red fluorescent protein.

Educational **Events**

Maternal and Child Health Symposium

Dr. Souhaila Al Khodor and Dr. Annalisa Terranegra opening the Maternal and Child Health Symposium

Sidra Medicine's first ever Maternal and Child Health Symposium welcomed nearly 600 delegates. The three-day event focused on the latest developments and innovations in maternal and child health, patient care and research.

In alignment with Qatar's National Health Strategy 2018-2022 priority targets: Healthy women leading to healthy pregnancies, the MCH Symposium 2020 showcased presentations and panel sessions from more than 32 local and international experts in maternal and child health. It was co-chaired by our researchers, Dr. Souhaila Al Khodor Director of Maternal and Child Health Department and Investigator in Nutritional Genomics and Metabolism.

The three-day symposium was structured on the three biological transitions that a woman's body goes through during pregnancy: pre-conception; pregnancy and fetal; and postnatal. Sessions covered topics related to understanding the role of lifestyle and women's health in fertility; the role of infectious and chronic diseases in reproductive health as well as the latest updates on the novel tools and protocols used in IVF and in management of pregnancy complications.
The conference also marked International Women's Day with the delegates on 8th March.
Dr. Khalid Fakhro, Acting Chief Research Officer said: "MCH 2020 showcased the successful integration of our clinical and research maternal programs at Sidra Medicine. International and local speakers shared their experience on topics such as gestational diabetes, pre-term birth, perinatal child health and reproductive conditions such as infertility that affect millions of women and men worldwide."

During a session presented by Prof. Kent L. Thornburg, titled "The Developmental Origins of Health and Disease paradigm: Maternal Roots of Adult Onset Chronic Disease", it was noted that the introduction of good nutrition in the early stages of a woman's life is fundamental not only for her health, but also for healthier pregnancies and healthier babies. The theory was supported by several speakers during MCH 2020, who shared best practice examples and research programs from Qatar as well as from the United States, the United Kingdom, Spain and other countries.

Dr. Souhaila Al Khodor said: "Our research theme for MCH 2020 was built around the philosophy that health in the early years of childhood is the foundation of health throughout the life course. By bringing together both local and global experts (in person and remotely via videoconference), it was enlightening to see that we share a common goal – and that is, in order to build a healthier society, we must focus on improving maternal and child health."

Dr. Annalisa Terranegra, Principal Investigator, Assistant Level, Laboratory of Nutritional Genomics and Metabolism said: "Our work at Sidra Medicine, in particularly in partnership with entities like the Ministry of Public Health and Hamad Medical Corporation - puts us in a unique position to collaboratively develop and implement strategies and healthcare approaches that can directly improve maternal and child health outcomes for families in Qatar."

MCH 2020 wrapped up on the 9th of March, with key calls to action on better nutrition for women, importance of HPV vaccines in adolescent women, and investment in further research to understand pregnancy complications.

Educational **Events**

Precision Medicine and Functional Genomics 2020

We hosted our fifth annual functional genomics symposium virtually this year. The theme — "Precision Medicine and Functional Genomics 2020" (PMFG 2020) focused on key sessions related to the implementation of precision medicine solutions to advance patient care and the readiness to tackle pandemics such as COVID-19.

Over 50 renowned speakers from the research, clinical, biotechnology fields spoke at PMFG 2020; which attracted an online audience of more than 250 local and international scientists, researchers and medical professionals. Dr. Khalid Fakhro, Chief Research Officer said: "I would like to take the opportunity to thank everyone from our speakers, attendees, partners as well as sponsors and organizers who made our first ever virtual event a success. It was a privilege for us to connect with a global network of peers and colleagues who are all committed to finding innovative research-driven solutions that will transform patient care as well as address dynamic real-world problems like the current COVID-19 pandemic." Highlights of the symposium included keynote speaker, Professor Sir Mark Caulfield, Chief Scientist at Genomics England, discussing the 100,000 Genomes Project and the impact of such large scale discoveries on transforming patient care. Shedding light on the collaboration between Qatar Genome Programme, Qatar Biobank, Sidra Medicine and Weill Cornell Medicine Qatar regarding Qatar's first Gene Chip (Q-Chip), Professor Ronald G. Crystal, Chairman of the Department of Genetic Medicine at Weill Cornell Medicine in New York explained how the project allows for the rapid identification of individuals who are carriers for Mendelian (genetic) diseases in the population. Such identification methods are a key part of Qatar's Precision Medicine Program, as they allow to significantly improve the genetic diagnosis of a large number of diseases. Unlike the genetic results that come from international laboratories abroad or gene assays developed using data from other populations, diagnosis using the Q-Chip is based on genetic information derived from Qataris, making the results obtained more relevant and accurate. It will also allow for families to obtain genetic counseling and make more informed decisions regarding their health and wellbeing and for family planning. Continuing on the session on precision medicine implementation programs, Professor Stephen Scherer, Director of the McLaughlin Centre and the Centre for Applied Genomics, The Hospital for Sick Children (SickKids) in the University of Toronto, Canada, discussed how precision medicine and the science of genomes is unlocking the secrets behind Autism.

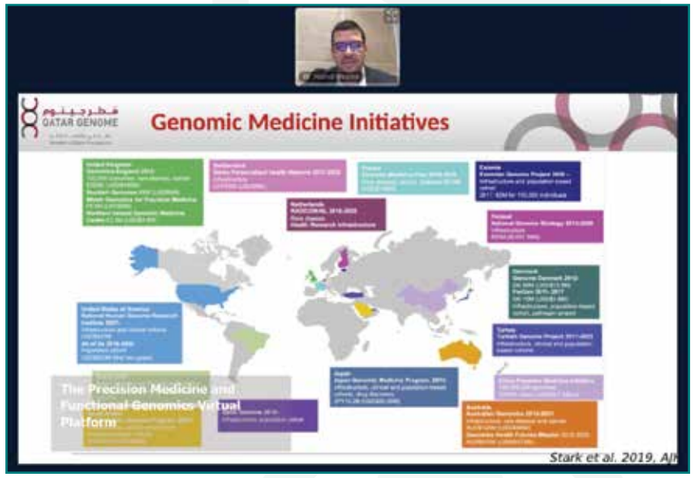
Planning Committee of the Precision Medicine and Functional Genomics Virtual Conference 2020

Professor Scherer highlighted the ongoing collaboration of Qatar National Research Fund, Sidra Medicine and SickKids through the BARAKA study to sequence and characterize 1,000+ patients with Autism Spectrum Disorder in the coming year. Dr. Amel Hassan, Senior Attending Physician at Sidra Medicine said during her discussion on precision medicine transforming the lives of children with Primary Immunodeficiency: "Physicians resort to genetic testing when patients do not react well to classic treatments.

If results show a change in a specific gene causing the illness, we carry on studying the function of the gene causing the illness as well as the treatment using precision medicine.

This is a practice that will enable pediatricians to start the treatment earlier and reduce the suffering of not only the patients but their families as well. I am proud to work for a healthcare organization like Sidra Medicine that is building and developing that concept into its patient care and treatment programs in Qatar." PMFG 2020's COVID-19 session was a hot topic, highlighting the global response to the pandemic and vaccines. Professor Laith Abu Raddad, Professor of Infectious Disease Epidemiology from WCM-Q and Director of the World Health Organization (WHO) Collaborating Center for Disease Epidemiology Analytics in Qatar indicated that Qatar's proactive and swift response has prevented a second wave now for more than six months after the first wave and so far there is no sign of a second wave emerging. He elaborated that due to the successful containment of the virus as well as early public health planning and interventions, Qatar was in a strong position in being able to successfully protect its population from a second wave and is now looking into the future with ambitious plans for vaccination to emerge from the pandemic and return to normality. Dr. Khalid Fakhro continued: "The impact of Qatar's investment in research and healthcare really came to the forefront during the pandemic. Not only were we in a position that we could mobilize our research efforts to support the country's testing requirements at Sidra Medicine but we also developed our own new testing methods highlighting resource and cost efficient ways to manage large scale testing. This speaks to not only the readiness of the country in tackling pandemics like COVID-19 but also the level of expertise and commitment within the scientific community in advancing the national healthcare and precision medicine agenda." PMFG 2020 conference co-organizers

Dr. Damien Chaussabel and Dr. Bernice Lo from Sidra Medicine said: "PMFG 2020 announced 50 highly competitive abstract submissions at PMFG 2020, six of which were selected for oral presentations. Sidra Medicine is committed to encouraging a culture of education and growth and we would like to acknowledge the trainees who attended the symposium this year, and we hope that they found the experience educational and insightful."



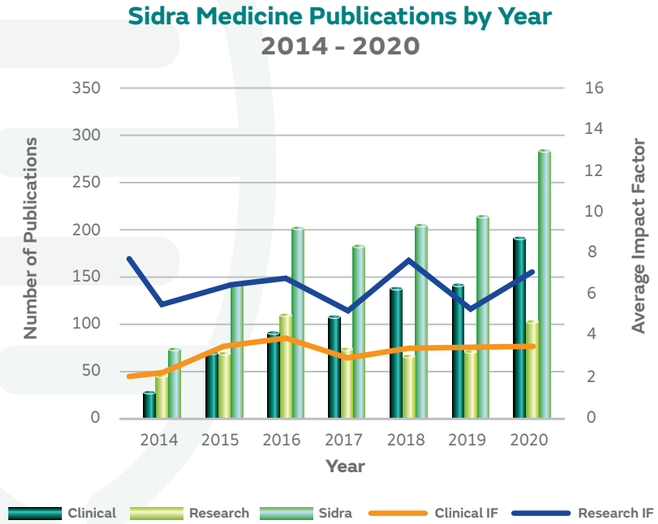
The Precision Medicine and Functional Genomics Virtual Platform

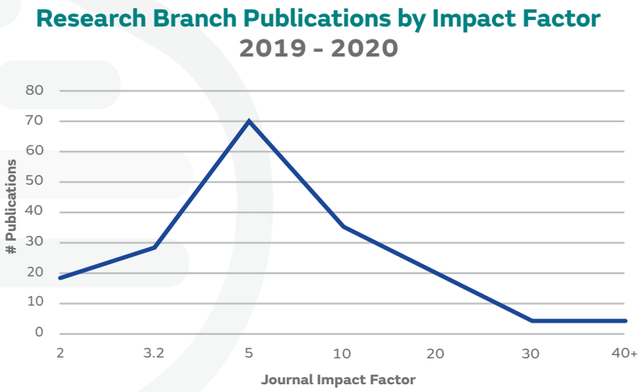
Publication Registry

Publication **Registry**

Publications

Scientific publications are an important measure of output and an integral outcome of Research. It serves to share with the world gained knowledge and expertise. At Sidra Medicine, we are an academic research hospital, where we have dedicated support to clinicians to perform research and disseminate their outcomes. We are proud to have a steady output of high-impact publications, with well above 70% of Research Branch publications being published in the top 15% of internationally-recognized biomedical journals.





Some of the key 2020 Research Branch Outcomes are summarized as follows:

- 105 publications with a mean impact factor of 7.1
- 74% of publications are in the top 15% of internationally-recognized journals
- 92 formal scientific collaborations locally and internationally
- Over 60% of PIs with academic affiliations inside Qatar
- Research Branch investigator average H-Index for 2020 is 21
- Over 20 MSc and PhD students were trained in the PI labs
- 46% of research studies have a clinical lead/co-investigator

Publication **Registry**

Genetics

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