

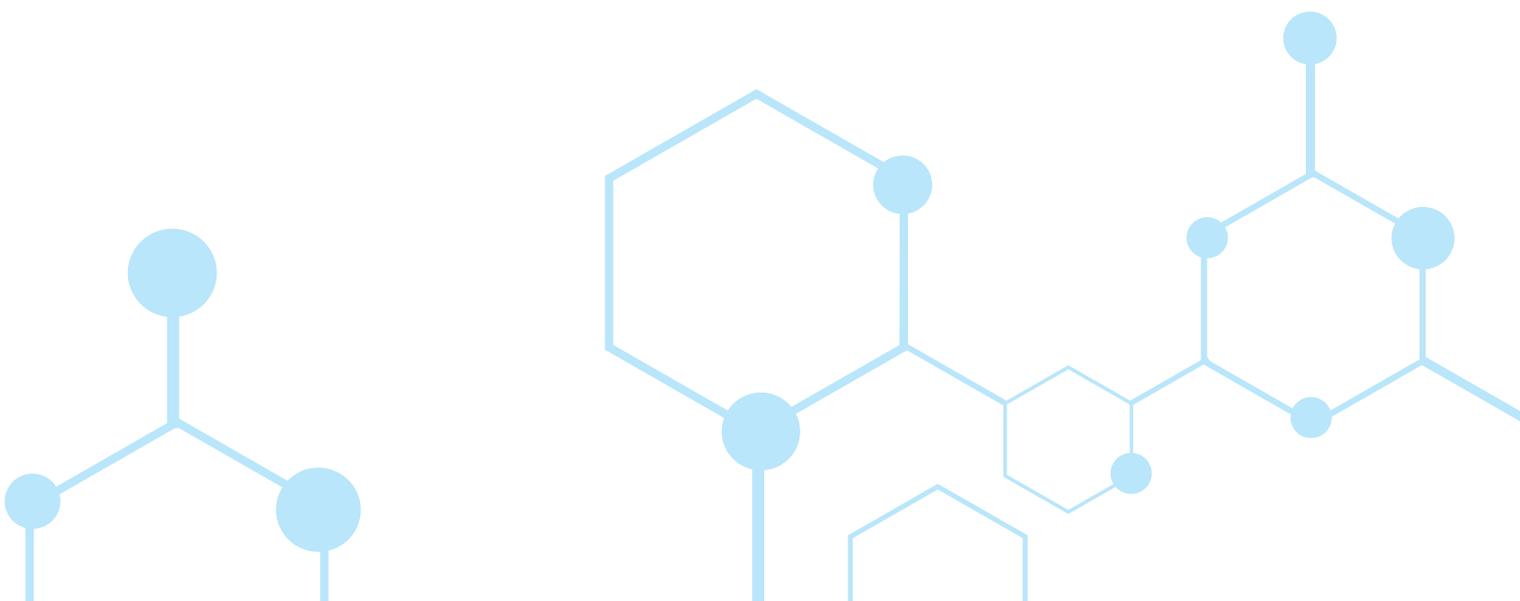


**SIDRA MEDICINE | RESEARCH**  
Annual Report 2021





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Annual Report 2021





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# WELCOME NOTE FROM THE CRO

In many ways 2021 has been a challenging year: the continued uncertainty surrounding the global pandemic, the burden on healthcare facilities, and the anxieties we all had to endure made adapting to a new way of life quite 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 our strength as a national academic medical center, the Sidra Research Branch made significant strides during the pandemic. On one end, our Core Genome Laboratory developed methods to detect viral infections that cleverly bypassed global shortages for testing reagents. On another, our scientists joined global consortia investigating penetrance and variability of COVID-19 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 Corona viruses. Even outside of COVID-19 projects, the Research Branch had a remarkably successful year. Reaching a milestone of 6,000 patients and family members enrolled in research studies. Sidra clinicians and scientists published nearly 300 papers, with almost 85% of Research Branch publications in the top 15% of international journals. Importantly, Sidra Medicine was awarded 12 national grants totaling QR 7.3M, demonstrating the growing recognition of the importance of academic medicine for translational discovery.

In terms of contribution to Qatar's knowledge economy, Sidra continued its mission 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 52 trainees and students were part of the Sidra Research family in 2021, including almost 20 MSc & PhD Students, 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 building a research enterprise in Qatar with strong links to the global scientific community.



In summary, 2021 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 that discovery and innovation remained strong, to our diverse clinical collaborators who were fundamental in devising studies and translating outcomes to patients, and, most importantly, to all the families and patients who entrust Sidra Medicine to deliver the highest-quality, research-driven care.

**Dr. Khalid A. Fakhro**  
**Chief Research Officer**

# LEADERSHIP STRUCTURE



**Khalid Fakhro**  
Chief Research Officer



**Rashid Al Ali**  
Executive Director  
Research Core Facilities  
and Digital Health Core



**Max Renault**  
Director  
Research Operations  
and Services



**Damien Chaussabel**  
Executive Director  
Division of  
Translational Medicine

# RESEARCH MISSION AND STRATEGY

# 2021

## *Our Year of Big Thinking, Collaborations and Discovery*

### **Our Vision**

To establish a strong, clinically oriented biomedical research program, and to develop a national resource of genomic information that improves health in Qatar and the region.

### **Our Mission**

We will 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 Medicine.

### **Our Strategy**

- Patient Driven Research
- Advanced Diagnostics
- Personalized Therapy

These strategic pillars require close collaboration, trust and interactions between patients, trainees, physicians, and researchers within Sidra, as well as in Qatar and around the world to advance care at Sidra.

In summary, this endeavor will become a key differentiator for Sidra Medicine, setting it 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.

### **Our Values**



# INTERNAL RESEARCH COUNCIL

As part of the IRC responsibilities, the committee has been reviewing the scientific merit and alignment of all clinical trials submitted by both Research and Clinical Investigators and will continually and critically review existing research projects and evaluate their impact.

Developing a national resource for genomic information on the population of Qatar to further research that improves health in Qatar and the region has largely been established as Sidra is now the National Sequencing Core, that does Sequencing and Bioinformatics on large scale, primarily for the Qatar Genome Program and internally for patients.

The aim now is to set an agenda to establish a strong, clinically oriented biomedical research program with significant national relevance, therefore, the duty of the IRC to strengthen Clinical Research integration and make Sidra one of the best institutions in the world for Clinical Research.

The committee meets regularly, aiming to develop Sidra's translational research roadmap (Precision Medicine), maximize Clinical-Research integration, and streamline research projects for strategic alignment, funds utilization and impact.

As 2021 progressed, IRC established project review elements on which to critically review existing research projects and evaluate their impact.

The review elements cover the following:

- Evaluation of alignment with Sidra Research Strategy
- Achievements/Output
- Impact evaluation of the research or treatment on the health of individuals and populations.

## CHAIRPERSON

**Khalid A. Fakhro**

Chief Research Officer

## VICE CHAIRPERSON

**Ibrahim Janahi**

Division Chief

Pediatric Pulmonology

## MEMBER

**Damien Chaussabel**

Executive Director

Translational Medicine Department

## MEMBER

**Davide Bedognetti**

Director

Human Immunology Division

## MEMBER

**Khalid Hussain**

Division Chief

Endocrinology

## MEMBER

**Souhaila Al Khodor**

Director

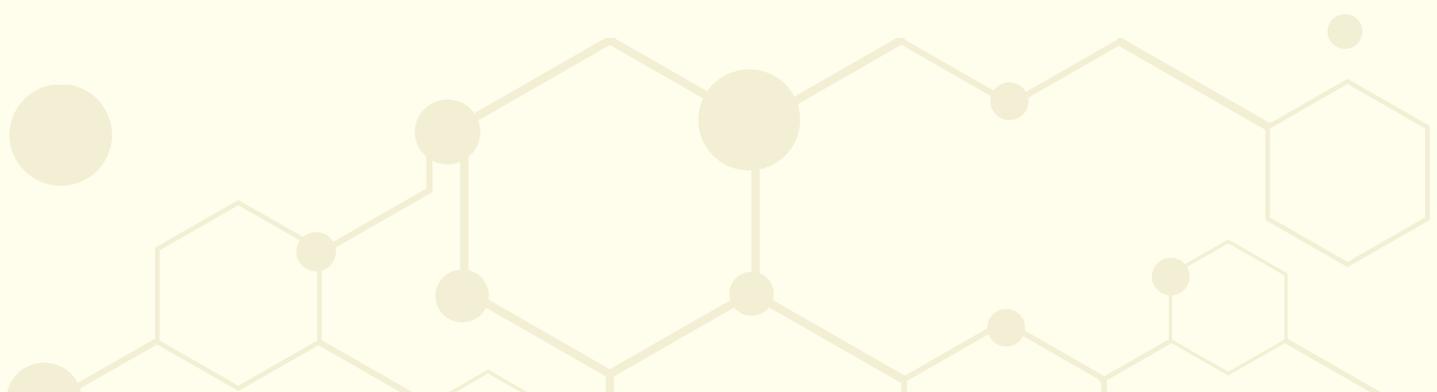
Maternal and Child Health Division

## MEMBER

**Colin Powell**

Senior Attending Physician

Emergency Department

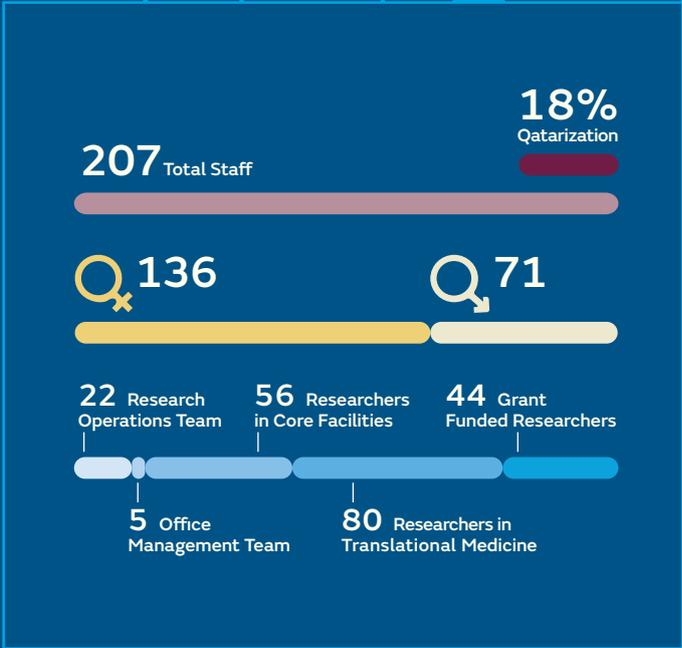


# SIDRA MEDICINE: OUR RESEARCH AT A GLANCE



## 36 Nationalities at Sidra Research

Algeria	Netherland	Jordan	Qatar
US	Egypt	Lebanon	KSA
Australia	Philippines	Macedonia	Somalia
Azerbaijan	France	Morocco	Spain
Bahrain	Germany	New Zealand	Sudan
Belgium	India	Oman	Syria
UK	Iran	Pakistan	Thailand
Canada	Iraq	Palestine	Tunisia
China	Italy	Portugal	Yemen



### Public and Community engagement & education



**69.5%** of Sidra Medicine Research staff have attended International and Local conferences, seminars and symposiums



More than **60%** of our Senior Staff hold Adjunct Faculty Appointments in Qatar and around the world



**103** Oral Presentations conducted around the world

# COLLABORATIONS

**104**

Total Collaborations

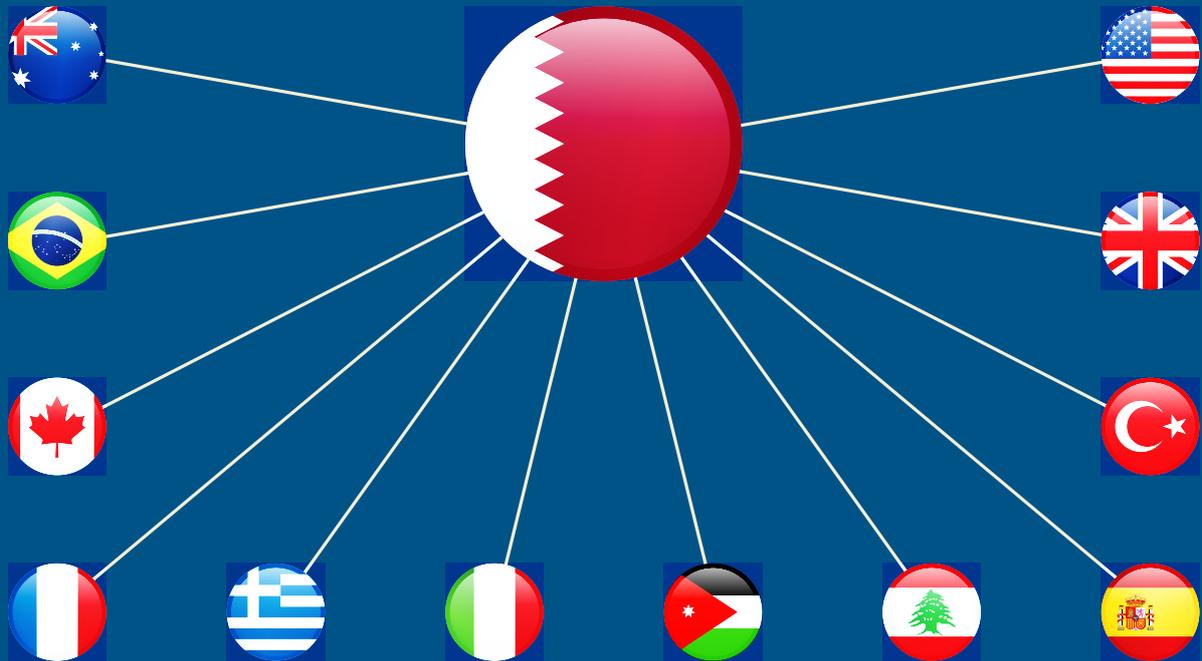
**75**

International Collaborations

**Countries we have international collaborations with:**

Australia  
Brazil  
Canada  
France  
Greece  
Italy

Jordan  
Lebanon  
Spain  
Turkey  
UK  
USA



**29**

Local Collaborations

**Local Institutions we collaborate with:**

Hamad Medical Corporation  
Hamad Bin Khalifa University  
Qatar University  
Texas A&M University at Qatar  
Qatar Museum Authority  
Weill Cornell Medicine – Qatar  
Anti-Doping Lab Qatar  
Equine Veterinary Medical Center

# PUBLICATIONS



**300+**  
Sidra-affiliated peer-reviewed scientific papers published in 2021



**122**  
Research Branch papers published in the last 12 months



**85%**  
of publications are in the top 15% of internationally recognized journals



**21**  
Research Branch investigator average H-Index for 2021



**47%**  
of research studies have a clinical lead/co-investigator



**6.9**  
Mean impact factor for Research Branch publications in 2021

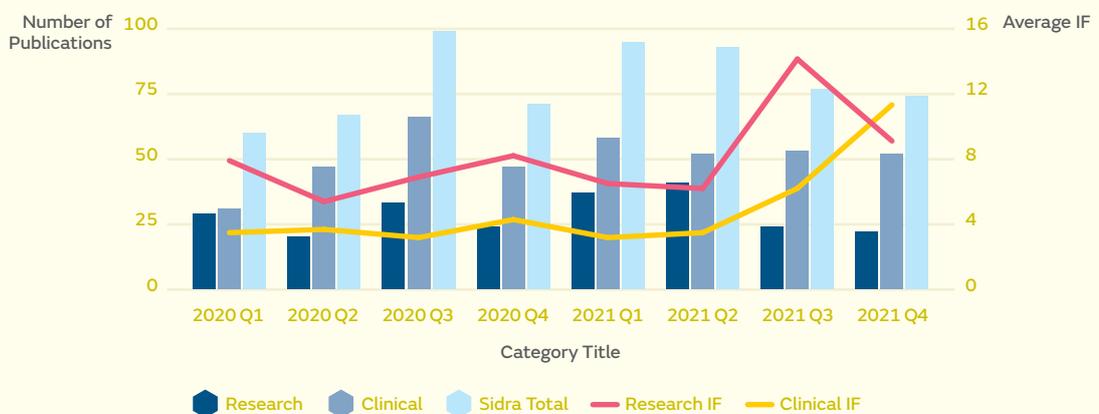


**104**  
feature in top 15% of journals worldwide



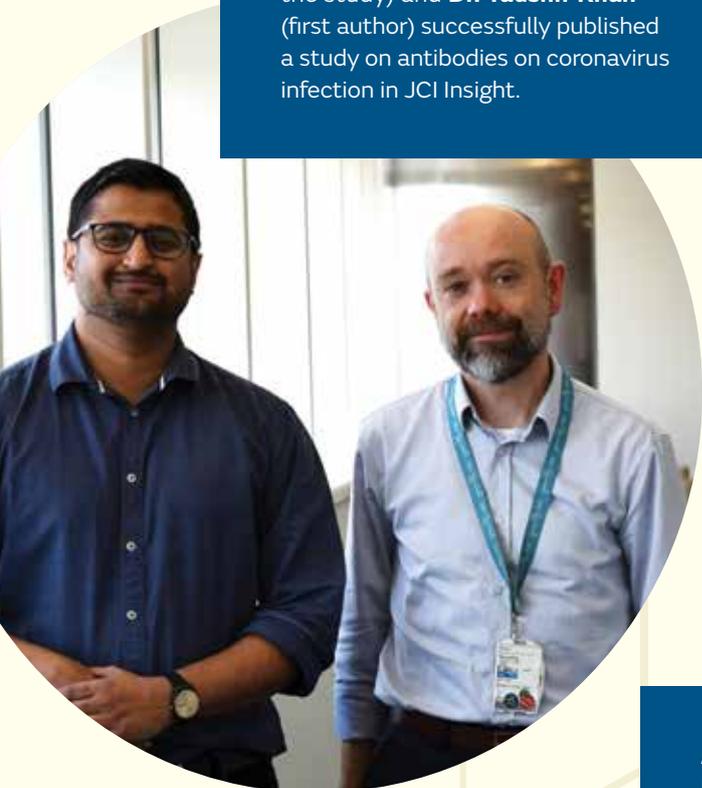
**22**  
articles in the top 2% of journals worldwide

## 2020–2021 Sidra Medicine Publications

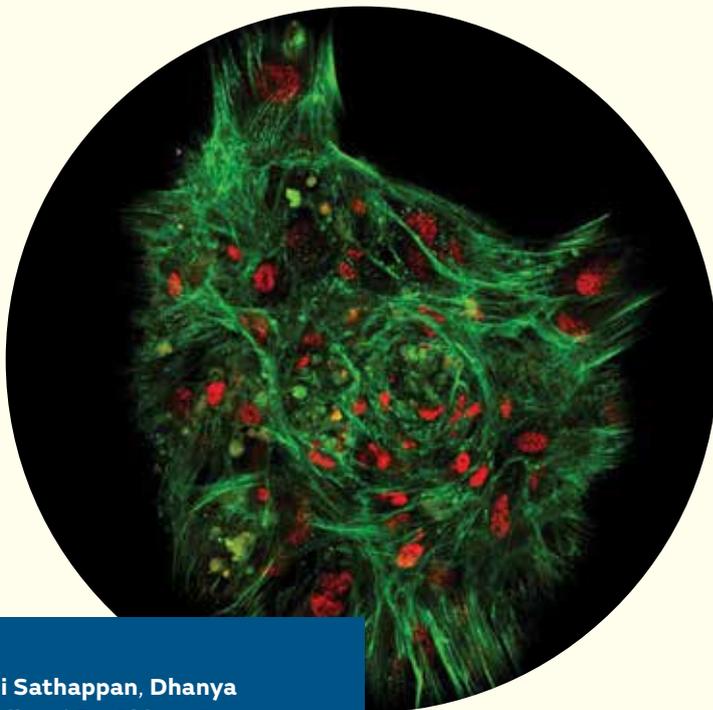


# AWARDS AND ACHIEVEMENTS

**Dr. Nico Marr** (Group leader of the study) and **Dr. Taushif Khan** (first author) successfully published a study on antibodies on coronavirus infection in JCI Insight.



**Abbirami Sathappan, Dhanya Kizhakayil** and **Dr. Chiara Cugno** were awarded the 1st place in the 5th BRIO Cycle (Best Representation Image of an Outcome) for their image titled "A Cell Bouquet."



The article "Toward Nanotechnology-Enabled Approaches against the COVID-19 Pandemic" by **Dr. Davide Bedognetti** and **Dr. Laura Fusco** was voted in 2021 by the nanoscience community as 2020's top ACS Nano article. It received 15,983 votes out of a total 35,878.



## BREAKOUT STORY

# Research Team at Sidra Medicine Studying the Potential of Precision Medicine for Acute Asthma in Children



Asthma is one of the most common childhood illnesses in the world. An acute attack of asthma requiring a trip to the hospital may have a huge impact on the child and their family. Some children respond much better than others to the currently available standard treatments for acute asthma. An ongoing study at Sidra Medicine may explain why this happens, thereby applying personalized asthma treatment by investigating the genetic and cellular background of acute asthma in each individual child.

At Sidra Medicine, **Professor Colin Powell**, senior attending physician, and **Dr. Nicholas van Panhuys**, head of the Laboratory of Immunoregulation, have joined the forces of clinical and research expertise and technology to study the relationship between a person's genetic background and their response to treatment with asthma medications used during an acute attack.

Precise and effective management of asthma is particularly important in children as studies around the world have recognized asthma as the leading reason behind chronic absenteeism in school, ability to participate equally in sports and exercise and has an impact on sleep, quality of life of child and the family.

“By studying the genetic background of acute asthma attacks, we may be able to classify acute asthma more precisely and employ individualized treatment through Precision Medicine,”

said Dr. Powell. “There are many studies examining the genetic background in children with chronic asthma and how they respond to treatment, but there are fewer studies on acute attacks, so that’s the exciting novel territory that we’re stepping into,” added Dr. van Panhuys. One aspect of the ongoing project will also be to explore asthma genes that are particularly relevant to the Qatari population.

The team is developing an array of genes and recording the associated responses to common asthma medications based on the treatment provided to children in the ED. As a result, future pediatric patients with acute asthma at Sidra Medicine may be given a tailored medication regime based on their individual genetic makeup, which is a great example of Precision Medicine. Not only could this streamline management of the patients by giving them more personalized treatment, but it will also reduce the worry of parents who sometimes find even the most common asthma medications to be ineffective on their child.

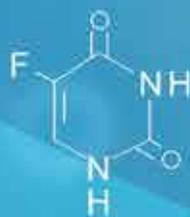
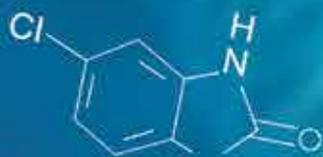
The close collaboration between the clinical research expertise of the ED and the genomics laboratories of Sidra Medicine is making the study holistic and thus paving the way towards novel treatment approaches for acute asthma in children around the world.

**Written by Areesha Lodhi**





RESEARCH  
DIVISIONS



# SCIENTIFIC DIVISIONS

## TRANSLATIONAL MEDICINE

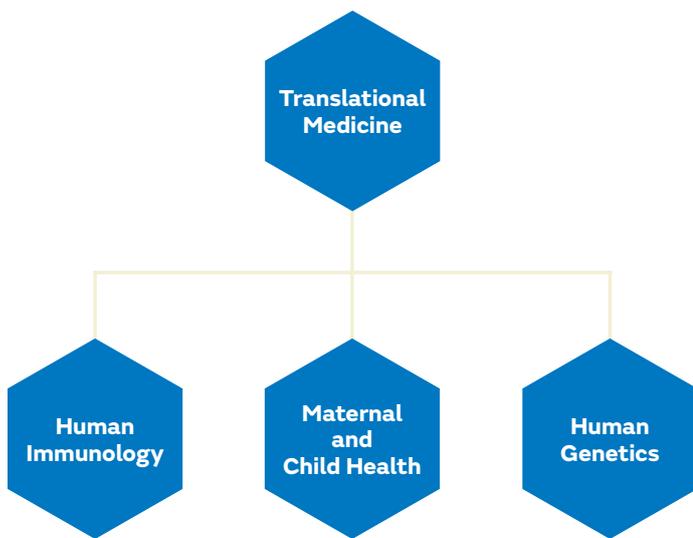
The Translational Medicine department is a key enabler for Sidra Research's strategy, which will lead to the establishment of Sidra Medicine as a world-class academic medical center and a destination for patients seeking the best available care in the region. The translational medicine department will focus on the development and implementation of precision medicine approaches. The Department of Translation Medicine is divided into three divisions, where all research groups fall under. As a result of the commitment of Sidra to innovation in the field of precision medicine, the research groups at Sidra are engaged in research activities leveraging high throughput profiling technologies in the context of patient-based research.



### EXECUTIVE DIRECTOR

## Dr. Damien Chaussabel

Dr. Chaussabel obtained his PhD from the University of Brussels in 1999. As a trained immunologist, he acquired expertise in the genomics and bioinformatics fields, with the use of whole genome transcriptional profiling tools as a postdoctoral fellow at the NIAID/NIH for the study of host-pathogen interactions. Prior to joining Sidra, Dr. Chaussabel developed a genomics and bioinformatics program at the Baylor Institute for Immunology Research in Dallas TX (2004–2010). He served as head of the Systems Immunology Division at the Benaroya research institute in Seattle WA (2010–2014) where he led studies investigating “genomic reprogramming” that occurs in the blood of patients with infectious and autoimmune diseases as well as in response to vaccination.



# Human Immunology



**SCIENTIFIC DIVISION DIRECTOR**  
**Dr. Davide Bedognetti**

Dr. Davide Bedognetti, MD, PhD is the Director of Cancer Program at the Sidra Medicine Research Branch and serves as Adjunct Associate Professor at the Hamad Bin Khalifa University in Doha, Qatar. Dr. Bedognetti joined Sidra in 2014. He received his MD and PhD in Clinical and Experimental Oncology and Hematology from the University of Genoa, Italy.

From 2013 to 2014, he 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. He is the Editor of the Cancer Microenvironment Section of the Journal of Translational Medicine (Springer Nature), and Editorial Board Member of Scientific Reports (Nature Research) and Cancer Treatment Reviews (Elsevier).

Dysregulation in the immune system can cause or favor a wide spectrum of human illnesses that are prevalent in women and children, ranging from classical immune-mediated diseases (e.g, asthma, autoimmune diseases, and immune deficiencies) to multi-factorial pathologic conditions such as cancer, infections, neurological disorders including autism, complicated pregnancy, and pre-term birth. The availability of therapeutic approaches aiming at reprogramming the immune system are constantly increasing, and with them the need to tailor treatment choices, predict response and manage and/or anticipate adverse events. By studying the immune system, we can improve efficacy of vaccination, repress autoimmunity or reverse conditions such as cancer and infections. The division is divided in two sections, which are (a) The Allergy, Inflammation and Infectious Disease (AIID) Section, which focuses on infectious disease, inborn errors of immunity, asthma, allergy, and complicated pregnancies, and (b) The Cancer Immunology and Immunotherapy (CII) Section, which focuses on cancers in women and children. The mission of the division is to to develop novel diagnostic approaches and therapeutic strategies to promote and realize the concept of precision medicine in patients affected by diseases caused or facilitated by immunologic dysfunctions. The division will serve as a beacon for the training new generation of translational scientists with knowledge of both basic and clinical aspect of human immunology.





## Maternal and Child Health

A woman's health at conception and during pregnancy impacts the wellbeing of her child. Sidra's Maternal and Child Health (MCH) division 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 MCH 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.



### SCIENTIFIC DIVISION DIRECTOR **Dr. Souhaila Al Khodor**

Dr. Al Khodor is the Director of Maternal and Child Health Department in the Research Branch at Sidra Medicine, Qatar, since July 2019 and an Investigator-associate level since January 2015. Dr. Al Khodor is in charge of the Microbiome and Biomarkers discovery lab. 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). Before joining Sidra, Dr. Al Khodor worked 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. Dr. Al Khodor is an adjunct Assistant Professor at the College of Health & Life Science in Hamad Bin Khalifa University, and an Adjunct Assistant Professor at the Department of Biomedical Sciences, College of Health Sciences in Qatar University.



## PROGRAM HIGHLIGHT

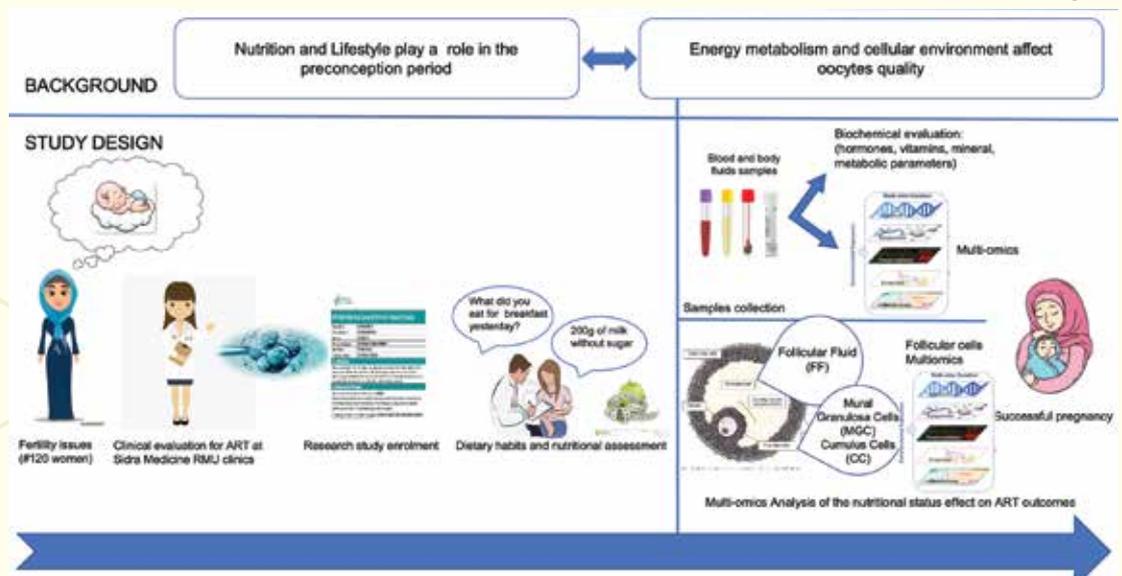
### Two new studies successfully launched

MCH department recently launched two new studies addressing the major focuses of the program: the “Omouma” and “PN-ART” studies, both fruits of the successful collaboration between MCH research team and Women’s Services at Sidra Medicine. “Omouma” means motherhood in Arabic, implicitly explains the aim of the study, which is to investigate factors affecting pregnancy outcomes, the early life determinants and their impact on infant, child’s and adolescent’s health in the population of Qatar. We will monitor the pregnant women every trimester until delivery and one-year post-partum. Using the biological samples collected from the mother-baby pairs, we aim to identify the molecular mechanisms of pregnancy complications and to identify biomarkers that can help predicting pregnancy outcomes and may pave the way for a personalized intervention. We will also investigate the maternal, genetic, social, environmental, lifestyle on the infant’s health and how early childhood determinants affects health outcomes of the infant, child and adolescent. This study is a great example of a multidisciplinary effort including research team, women’s services and psychiatry departments at Sidra Medicine.

According to a report by the World Health Organization published in 2010, fertility rates in all six Gulf countries fell significantly between 1980 and 2005. In Qatar, the fertility

rate per woman dropped from 6.1 children to less than two in the last 50 years. The role of environmental factors, such diet, lifestyle, and microbiome on the preconception period has already been established. However, there is still a knowledge gap in understanding the molecular mechanisms governing the interaction between diet and the follicular metabolism and its effect on the outcomes of the assisted reproductive technology (ART). We planned an observational study named “Precision nutrition in Assisted Reproductive Technologies (PN-ART).” The study will enroll women from the Reproductive Medicine unit with the aim of exploring the association between women’s nutritional status, their blood and follicular molecular signature, in addition to the gut and vaginal microbiota composition, and their ART outcomes. We will recruit the women before starting any fertility intervention, collecting information on nutritional status, dietary habits, medical history along with a set of samples for multi-omics applications. We will also follow up the women during the fertility procedures, collecting information on the ART outcomes. Understanding how the nutritional status and diet profile of women affects their follicular function may open a new pathway to improve the efficacy of ART procedures. This study is the result of a successful collaboration between Sidra Medicine MCH team and the Reproductive Medicine unit.

#### Diet effect on the Follicular Cells Metabolism: toward Personalized Nutrition in Assisted Reproductive Technologies



## Human Genetics

The Human Genetics Division at Sidra Medicine Research Branch harbors basic and translational multidisciplinary research in broad areas of genetics, molecular genetics, genomics and bioinformatics, with the goal of enabling the institution's mission to deliver world class tertiary care for women and children. The division's overall strategy is to leverage state of the art techniques in collaboration with local and world's experts to drive the precision medicine of a wide spectrum of genetic disorders afflicting the population of Qatar and of the wider Middle East. The mission of the Human Genetics Division at Sidra is to apply multidisciplinary expertise to further our understanding of how human genetic variation impacts health and disease in women and children. It's ultimate goal is to help bring the highest quality of care possible to patients with genetic and genomic disorders, ranging from precise diagnostics and risk profiling to state of the art therapeutics. The division's core contribution is to enable patients access to cutting-edge research not yet implemented at the clinic, having the promise to shedding light on their diseases and bringing hope for new treatment options.



### SCIENTIFIC DIVISION DIRECTOR **Dr. Khalid Fakhro**

Dr. Khalid Fakhro is the Chief of Research and Director of the Precision Medicine Program at Sidra Medicine, the largest tertiary care women and children hospital in Qatar. Dr. Fakhro leads the Laboratory of Genomic Medicine, which focuses on bringing emerging genomic technologies from the lab close to the patient's bedside. Over the past decade, his group has sequenced thousands of genomes from patients and volunteers across the Middle East, leading gene discovery efforts for a wide range of rare disorders, as well as landmark studies on population structure, genome structural variation, and the role of Islamic ethics in genome research. In addition to research and hospital duties, Dr. Fakhro serves multiple leadership roles in Qatar's growing biomedical ecosystem, including as Board Member of the Qatar Precision Medicine Institute, and Adjunct Faculty at both Weill-Cornell Medical College and Hamad Bin Khalifa University.

## PROGRAM HIGHLIGHT

### Qatar genomics study unlocks secrets to shaping future of healthcare for millions

Sidra Medicine Researchers have unveiled a high resolution map of the genetic structure of Arab and Middle Eastern populations, providing new insights into human history in the region and ancestral patterns that may help to explain local human traits and disease risks.

At present the Qatar study—published in the leading scientific journal *Nature Communications*—reveals that ancient populations in the Arabian Peninsula played a far more central role in the story of early human migration out of Africa than was previously understood.

Developed by an international team led by Dr. Younes Mokrab and Dr. Khalid Fakhro from Sidra Medicine in Qatar, in collaboration with Qatar Genome Programme, it is the first large-scale analysis of the genetics of Arab and Middle Eastern populations. DNA from more than 6,000 people living in Qatar has been examined, with their genomes compared to those from other populations living around the world today, as well as ancient DNA.

Understanding the genetics of these under characterized populations breaks down a barrier to

precision medicine tailored to address disease risks unique to people with Middle Eastern ancestry.

The study revealed key novel historical and social insights into Arab populations: A population split from early Africans occurred around 90,000 years ago, followed by a further split between 30–42,000 years ago that gave rise to the ancestors of modernday Arab, European, and South Asian populations. This is supported by the observation that Neanderthal DNA is far rarer in Arab populations than in populations that later mixed with ancient hominins.

The study found very high rates of homozygosity, which is likely to be a result of the tribal nature of Arab cultures, suggesting the suitability of this population in discovering novel disease risk genes and natural human knockouts.

Professor Asma Al Thani of Qatar Genome Programme stated: “As producers of the largest genomic dataset in the region, we hold a responsibility as Qatar Genome Programme to represent our part of the world and fill many of the existing knowledge gaps on genomics of the Middle Eastern populations. This paper is a great example of the role that we play.”

# CORE FACILITIES

## RESEARCH CORE FACILITIES

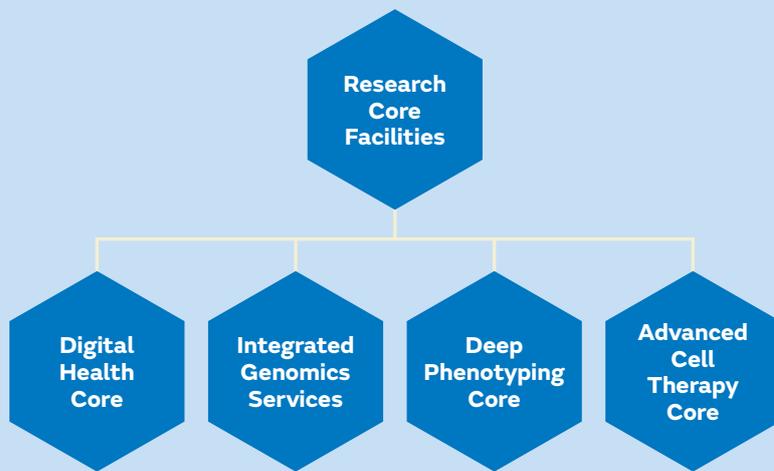
The Research Core Facilities support Sidra's mission to deliver Precision Medicine through Precise Diagnostics and Precise Therapeutics, unlocking the potential to leverage new knowledge through translational therapies and technologies intended to advance patients' care and safety through high quality and carefully supervised clinical trials.



### EXECUTIVE DIRECTOR Dr. Rashid Al-Ali

Dr. Rashid Al-Ali received his Ph.D. in Computer Science from Cardiff University – Wales, the UK in 2005 and his MS in Computer Science from George Washington University – Washington, DC, the USA in 1997. In 1992 Dr. Rashid Al-Ali graduated with a BS in Computer Engineering (with Honours) from the University of the Pacific – California, USA. Dr. Rashid Al-Ali was a Clinical Informatics Research Fellow at the Division of Clinical Informatics, Harvard Medical Faculty Physicians at BIDMC – Harvard Medical School, Boston USA in 2012.

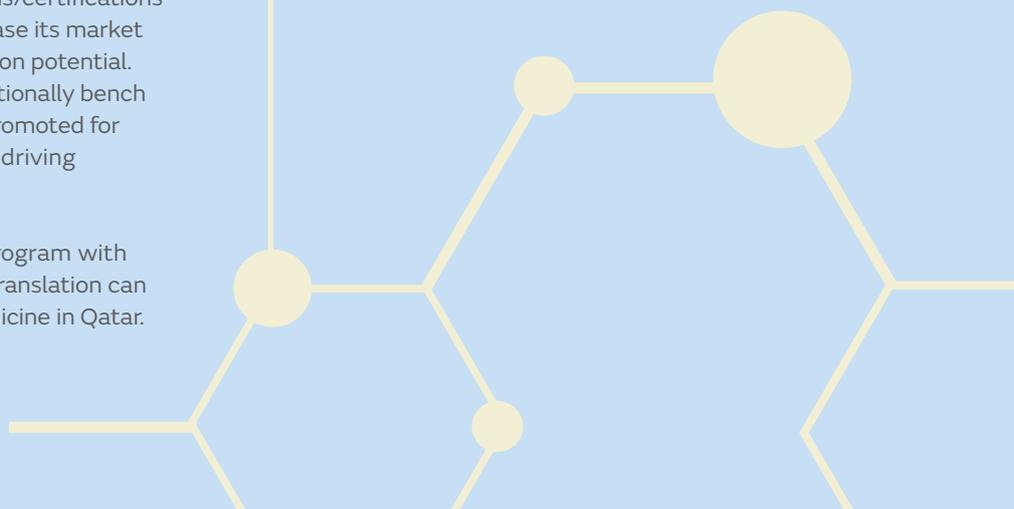
Dr. Rashid Al Ali is the Executive Director for Core Facilities and the Director of the Digital Health. The research core facilities consist of 4 cores: Deep Phenotyping, Advanced Cell Therapy, Integrated Genomics Services, and Digital Health. Dr. Rashid Al-Ali's research experience is in Distributed Systems, Grid Computing, and Clinical Informatics.



The philosophy of the Cores insists on 4 main leitmotivs:

1. Integration: functional crosstalk among the Cores to deliver.
2. Support of strategic program: all Cores' activities revolve around Precision Medicine, ranging from cutting-edge genotyping (Integrated Genomics Services (IGS)) and deep phenotyping (DPC) to the establishment of patients' registries and biorepositories (Biomedical Informatics Core) and the delivery of advanced therapies (Advanced Cell Therapy Core (ACTC)), both in experimental and/or defined clinical settings.
3. Accreditations/Certifications: services in each Core will be developed according to robust international standards ensuring highest quality of results/products, for the benefit of Sidra's patients. Accreditations/certifications will elevate Sidra's reputation and increase its market presence along with its commercialization potential.
4. Commercialization: high quality, internationally benchmarked services will be adequately promoted for both internal and external customers, driving downstream revenues.

A robust, high-quality clinical research program with integrated services and proactive clinical translation can be a significant differentiator for Sidra Medicine in Qatar.



## Digital Health Core

Technological advancements and biomedical device cost reductions resulted in the availability of a large amount of heterogeneous data that could provide insights and help advance precision medicine. To assist researchers in exploring data and answering research questions, it is critical to have an appropriate compute infrastructure and a talented multidisciplinary team to deal with data challenges.

The Digital Health Sciences division of Sidra Research aims to create a digital ecosystem to support all computational aspects (compute, storage, software development, data management, and analytics) critical to advancing precision medicine within Sidra and beyond. The division is critical in bridging the data exchange gap between the research and hospital departments.

The mission of Digital Health is to provide high-quality biomedical informatics services for all research projects by providing bioinformatics, compute, and storage infrastructure, assisting researchers and physician-scientists with tools/software, and assisting them in data management.

The Digital Health Department is more than just a data custodian; we thrive on making useful data available for consumption.

**CORE FACILITY DIRECTOR**  
**Dr. Rashid Al-Ali**



## CORE HIGHLIGHTS

### Sidra Research Biorepository

The biorepository platform is interdisciplinary project between various cores to establish and maintain a repository of biorepository data for the leftover samples from Sidra patients, routine procedures samples (RPS) and research ordered samples (ROS) to be used for future research as required.

The digital health department will develop the platforms required to aid in the method of providing researchers with access to biorepository resources for future research, to unify sample and data collection, storage, and processing, and to provide a searchable database to enable precision health.

### Research identification system (RIS-ID)

A major challenge facing the Research environment at Sidra is the lack of a unified method to identify an enrolled participant. Although each participant can be identified at the hospital, the compliance requirements of PHI often restrict Research environment from having access to these identifiers.

Storing the MRN number or having an application that keys in the MRN generally available in the Research environment is restricted. Although, this does not prevent individual researcher with IRB approval having access and store MRN as part of their research activities.

RIS-ID will create a way to generate a hashed identifier for each participant, which would guarantee uniqueness and uniformity in ID generation. These ID's can be referred later to check if the participant has already been enrolled for any study earlier.

### Interactive Variant Analysis (IVA)

The standalone desktop applications allow researchers to access their data stored on the HPC directly from their desktop, eliminating the need for additional settings or support.

Interactive Variant Analysis (IVA) which facilitates the filtering, analysis and interpretation of whole genome variant data. This interactive tool allows the identification of genes affected by deleterious variants that segregate along family pedigrees, case-control or sporadic samples.

Some of the features of IVA:

- Allow to load VCF files and samples together with clinical data
- High-performance and scalable VCF and gVCF indexing
- VCF normalization and variant annotation
- Clinical interpretation analysis of samples and families

## Deep Phenotyping Core

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 (DPC) 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 Medicine's Research Personalized Medicine agenda: "Establishing an Advanced Diagnostics program."



**CORE FACILITY DIRECTOR**  
**Dr. Jean-Charles Grivel**

Jean-Charles Grivel obtained his Ph.D. in immunology from the University of Aix Marseille II. He held several positions at the USA National Institutes of Health until joining Sidra in 2015. 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. Dr. Grivel has developed flow-cytometric methods for characterizing antigen-specific cellular responses as well as microvesicles and viruses. Dr. Grivel has received the NIH Award of Merit in 2006. He has authored 98 peer-reviewed publications. He currently directs the Deep Phenotyping Core of Sidra Medicine.



## 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 a state-of-the-art functional genomics Zebrafish Core Facility. 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.



### **CORE FACILITY DIRECTOR** **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. In 2018, he joined Sidra Medical and Research as Director of the Integrated Genomics Services, where he uses his background to enable more cost-effective, rapid and robust sequencing solutions, thus supporting important initiatives like the Qatar Precision Medicine Institute, but also enabling the use of cutting-edge sequencing technologies in a clinical setting for diagnostics and Precision Medicine. In this role, he is overseeing the activities of Genomics, Omics as well as the Applied Bioinformatics Core and the Zebrafish Core Facilities, and developing new platforms for large-scale biorepositories.

## CORE HIGHLIGHT

### Clinical Genomics Laboratory & Genomic Data Science Group Highlights



The global Covid-19 pandemic has posed great challenges for everyone, but most significantly impacted the healthcare and tourism industry. The continuous evolution of the virus poses a threat to overcome implemented countermeasures and requires the monitoring of populations not only for the presence of the Coronavirus, but also for the different variants in circulation. To support the national effort and working in close collaboration with colleagues from the Molecular Infectious Disease Laboratory, the CGL team has extracted and genotyped over 35,000 Covid-19 positive samples collected in the community and travellers. This effort enabled a rapid and measured adjustment of policy at the national level. More recently, CGL has deployed methods to perform whole-genome sequencing of SARS-CoV-2, which allows the detection of a wider range of variants that are hard to detect via conventional PCR methods, such as the new Omicron variant B.1.1.529.

The sequencing of human whole genomes and transcriptomes has developed into an indispensable tool for research and clinical diagnosis of rare and undiagnosed hereditary disorders and cancers. Until recently, these methods have been very costly to perform. With its most recent upgrade to Illumina Novaseq 6000 systems, a human whole genome can now be sequenced and analyzed for QR 2,300 in as little as a single day. This new sequencing technology is complemented by new approaches to generate sequencing libraries of higher quality using innovative high-throughput robotics like the recently upgraded Hamilton NGS Star and sptlabtech Mosquito, which enable more accurate sample handling at lower volumes. This allows to further reduce price, but also input material requirements, and lays the foundation for genomic Precision Medicine at scale.

## OMICS CORE FACILITY

### Rapid Advancement of Genomic Technologies

The Omics Core Facility is managed by Dr. Sara Tomei. The rapid advancement of the genomic technologies over the past decades has led to individualized diagnostic and therapeutic approaches, allowing the implementation of precision medicine. The Omics Core Facility works closely to the Clinical team to overcome challenges faced when working on archival tissue samples, such as formalin-fixed and paraffin-embedded (FFPE). FFPE samples

provide great insights on disease progression and prognosis. Yet, their histological assessment lacks reproducibility due to the subjective interpretation of the histopathologists. The molecular technologies implemented in the Omics Core Facility help overcoming such arbitrary histological interpretation, by providing reliable and robust methods for the digital counting of single molecules isolated from archival samples.

## ZEBRAFISH CORE FACILITY

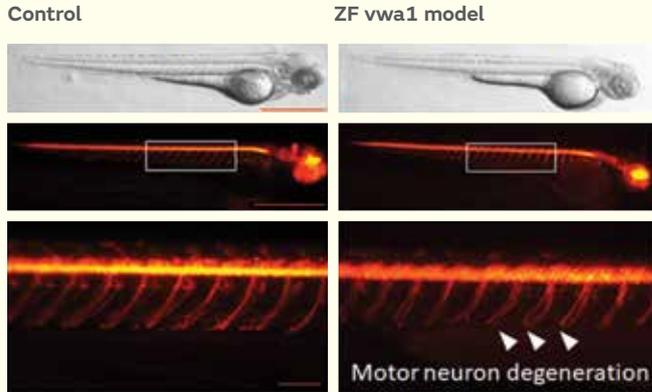
### Zebrafish Functional Genomic Core Facility, fishing for answers

The Zebrafish Core Facility is managed by Dr. Sahar Da'as. The Sidra Medicine Zebrafish Facility serves a significant role in developing clinically relevant zebrafish models of human genetic disorders and their related pathologies. The facility has developed a platform with the state-of-the-art equipment to study the genetic variation impact on cell, tissue, organ, and whole organism levels. The facility provides access to zebrafish model, techniques, and tools to utilize the model to its fullest potential. Zebrafish (*Danio rerio*), a small (3–5 cm) freshwater

tropical fish, offers the ability to accelerate genetic studies. It is an excellent vertebrate model for investigating human genetic diseases. The transparency of the zebrafish embryo during development has allowed researchers to track regulation of gene expression in real time in living animals. Comparison to the human genome shows that approximately 70% of human genes have at least one zebrafish orthologue. Out of the human genes of Online Mendelian Inheritance in Man (OMIM) database, (82%) of these genes can be related to zebrafish orthologous.

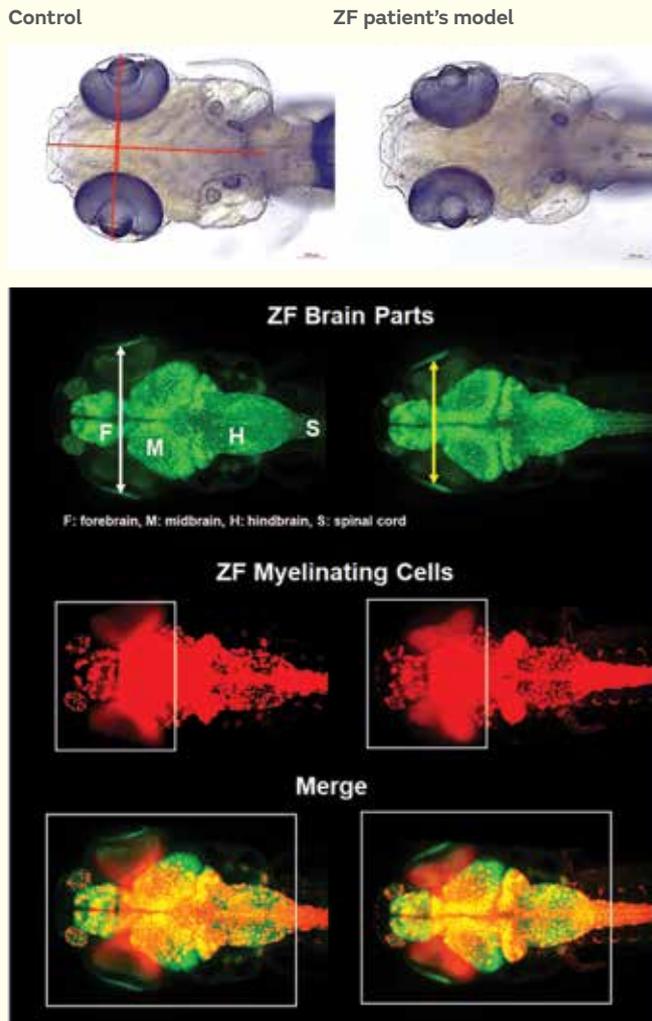
## ZEBRAFISH CORE FACILITY HIGHLIGHT

### Fish model links genetic variation to hereditary motor neuropathy



The facility team generated a model for a unique loss of function VWA1 gene mutation that was found in 17 individuals from 15 families in collaboration with UK scientists. The zebrafish vwa1 model demonstrated reductions in motor neuron axonal growth, synaptic formation in the skeletal muscles, and movement similar to patients' symptoms. This work was published in BRAIN journal, to read the full article: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8263055/pdf/awaa420.pdf>

### Fish model solves a child case with complex craniofacial anomalies



In collaboration with Dr. Fakhro's team and plastic surgery department, the facility team generated a zebrafish model for a 7-year-old patient presented to Sidra Medicine with craniosynostosis, dysmorphic facial features, eye anomalies, intellectual disability, and developmental delay. Zebrafish expressing the patient's unique gene mutation displayed a progressed microcephaly-like phenotype and head shape abnormalities. This work is submitted for publication.

## Advanced Cell Therapy Core



### CORE FACILITY DIRECTOR Dr. Chiara Cugno

Dr. Cugno joined Sidra in 2014, where she covers the roles of Director of the Advanced Cell Therapy Core and Attending Physician in Pediatric Oncology and Hematology. She is a 20-year experienced medical doctor with a Board Certification in Pediatrics and Pediatric Hematology/ Oncology at the University of Pavia (Italy), and a Master in Pediatric Hematology at the University “La Sapienza,” Rome (Italy). At Sidra, Dr. Cugno 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 a Clinical Trial Office.

The Advanced Cell Therapy Core (ACTC) synergistically completes Sidra Precision Medicine Program with its therapeutic component. ACTC’s mission and vision is completing the ideal pathway of the patient from advanced diagnosis to personalized treatment by:

- Supporting the Hematopoietic Stem Cell Transplantation (HSCT) Program which represents the cornerstone for the majority of other more advanced cell and gene therapy approaches
- Making Regenerative Medicine, Cell Therapy and Gene Therapy available to patients in Qatar and in the region
- Providing the management and coordination of clinical trials.

The delivery of cell therapy and gene therapy products entails a complex and highly articulated pathway starting with Research and Development and clinical research until scaling and production, all carried out in research and clinical laboratories aligned with National, British, US and European regulatory requirements and standards.

The Clinical Trial Office, recently established within ACTC, is striving to enable both Academic and Pharma-sponsored Clinical Trials, with benefits that range from strategic value—enhancing the hospital’s image to being a destination for the most advanced care—to quality value—improving the health of the local community—and finally to financial value—attracting new patients and preventing the outmigration of current patients.



## CORE HIGHLIGHTS

### Sidra Medicine Research develops eye drops to treat ultra-rare disease

Multidisciplinary team effort at Sidra Medicine produces country's first plasma eye drops, treats Qatari baby with "one-in-a-million" condition.

In early 2020, the family of a 10-month old Qatari child suffering from an ultra-rare disease called plasminogen deficiency, that was causing the child's eyes to fill with woody growths, got in touch with Sidra Medicine, a member of Qatar Foundation.

Doctors at Sidra Medicine, the only hospital in Qatar that offers precision medicine to pediatric patients, were positive that it could offer treatment for this condition. Dr. Chiara Cugno, Director of the Advanced Cell Therapy Core at Sidra Medicine, said: "When we were contacted about this case, we were confident right from the beginning that we had the expertise available to do this."

"The only thing was that the treatment—plasma eye drops—had never been produced in Qatar before, not just at Sidra Medicine, but anywhere else in the country. So, we understood that regulatory approvals would take a while, but since we already had a strong foundation to develop the eye drops in-house, we pooled in all our expertise to help this young patient."

The child's mother said: "There was a time my daughter couldn't open her eyes; I was terrified she was going to lose her eyesight."

The team working on producing the eye drops consisted of members from various departments across Sidra Medicine including Pediatric Hematology, Ophthalmology, Pathology Blood Bank and Advanced Cell Therapy Core.

In May 2021, the family received the locally produced eye drops for their child. Dr. Ayman Saleh, Chief of Pediatric Oncology and Hematology at Sidra Medicine said: "This was a truly joint effort, not just between the different teams at Sidra Medicine but also the family. We are grateful to the parents for trusting us and allowing us the opportunity to produce these eye drops. This project was not only a first for Sidra Medicine, but also a first for Qatar."

The child's mother said: "Having a top-notch hospital like Sidra Medicine tell us that they will make the medicine, store it, and ensure we have enough supply, makes me feel like we can sleep again, and that once again we have the time and energy for other things in life."

Despite the regulatory hurdles and the COVID related delays, the team is pleased they were eventually able to provide the child with the treatment.

## 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 ensures labs are in good working condition and practices safe.
- The Grants Office manages external and competitive awards.
- The Business Office handles budgets, financial reporting and procurement matters.
- The Outcomes & Reporting Office looks after strategic reporting, communications and outreach.
- The Governance & Compliance Office governs the conduct of all research and ensures compliance to relevant regulations and standards.
- 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.



### **CORE FACILITY DIRECTOR** **Max Renault**

The Research Operations and Services Core is led by Max Renault. Mr. Renault has 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 Management.

## CORE HIGHLIGHT

### Research Operations takes on Innovation & Tech Transfer role

An Office of Technology Transfer (OTT) is typically tasked with identifying, protecting, and commercializing inventions and related Intellectual Property (IP). IP can take the form of patents, trademarks, and trade secrets. For inventors to be adequately supported and for inventions to consistently be disclosed and benefits realized, a robust OTT office is crucial. In early 2021, the Research Operations team identified existing gaps and how to actively support inventors, in cooperation with the Legal department.

Research Operations mapped a process and established a partnership with Qatar Foundation's specialized OTT (known as the Industry Development and Knowledge Transfer 'IDKT'). As part of this relationship, QF-IDKT now facilitates active screening and processing of invention disclosures, ordering patentability/marketability assessments, and pursuing IP protection (provisional, full). Moreover, QF-IDKT with Research Operations will imminently support training and education needs of Sidra Medicine staff on the importance and risks of public disclosures, criteria for patentability, and more.

A very important outcome of capturing and managing IP is that inventions with promise can be advanced to commercial exploitation (e.g., through technology licensing, spinoff). This is of great benefit to not only Sidra Medicine but also to the inventor, who per institutional policy is entitled to a generous portion of any proceeds. For this to happen, potential commercial partners (e.g., big pharma, diagnostic test providers, resellers) must be identified and engaged. Research Operations is now looking to develop such capabilities internally and externally through strategic partnerships.

The pace of innovation can be extreme, especially in the biomedical arena, but ideas only in the mind of the inventor with no proper protection is of limited benefit. They must be expressed and implemented in practice before it is too late. You can contact [ResearchContracts@sidra.org](mailto:ResearchContracts@sidra.org) for more information and to assist you with your invention.







RESEARCH  
PRINCIPAL  
INVESTIGATORS

# RESEARCH PRINCIPAL INVESTIGATORS



**Dr. Ammira Akil**  
Precision Medicine  
in Diabetes Prevention

Dr. Akil is a principal investigator in human genetics program and the group leader of translational genomics of diabetes research team at Sidra Medicine research department. Dr. Akil has MSc in molecular Immunology, GC-LTHE1, Graduate Certificate in Learning and Teaching (Higher Education), PhD in molecular genetics from university of New South Wales, Australia. Dr. Akil earned an International Executive MBA from HEC – Paris business school focusing on “innovative management and Entrepreneurial leadership.” Dr. Akil has been a lead PI and Co-PI on national and international competitive research grant applications to support her diabetes research. 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. Dr. Akil scientific, organizational and communication skills, leadership, and management expertise in the field of clinical research placed her as the right person to found and chair the CUDOS nationally and internationally recognized scientific and educational series.



**Dr. Annalisa Terranegra**  
Laboratory of Precision Nutrition –  
Mother and Child Health Division

Dr. Terranegra obtained her MSc degree in Biological Sciences cum laudae in 2000 at the University of Siena, Italy, PhD in Molecular Medicine in 2007 at the University of Milan, Italy and Post-graduate Diploma cum laudae in Nutritional Sciences at the University of Milan, Italy in 2015. Dr. Terranegra covered a consultant position in genetics at San Raffaele Hospital, Milan, Italy (2010–2012) and a research fellow position in nutrition at San Paolo Hospital, Milan, Italy (2013–2014). Dr. Terranegra also has extensive teaching experience as Assistant Professor in Nephrology (2007–2010) and in Dietetic Sciences and Technologies (2010–2013) at University of Milan, Italy; Lecturer in Molecular Medicine PhD course (2009–2011) at University of Milan, Italy. Dr. Terranegra currently covers the position of adjunct Assistant Professor, since 2015, in the college of Health and Life Sciences at Hamad bin Khalifa University, Qatar and adjunct Assistant Professor, since 2018, in the College of Health Sciences at Qatar University, Qatar.



**Dr. Bernice Lo**  
Laboratory of  
Immunoregulation

Dr. Bernice Lo is a Principal Investigator in the Precision Medicine program at Sidra Medicine and an Adjunct Assistant Professor at Hamad bin Khalifa University (Doha, Qatar). She has contributed to the discovery, diagnosis, and molecular understanding of inherited autoimmune disorders. Bernice performed her post-doctoral training under the leadership of Dr. Michael Lenardo 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 under the mentorship of Dr. Jo Rae Wright, where she began her appreciation for the immune system and the critical role of immune tolerance and regulation.



### **Dr. Cristina Maccalli**

**Laboratory of Immune  
Biological Therapy**

Dr. Cristina Maccalli is a Principal Investigator at the Laboratory of Immune Biological Therapy, Translational Medicine Department, Research Branch, Sidra Medicine. Her expertise is in the field of immunology, tumor immunology and immunotherapy. She obtained a Ph.D. in Applied Genetics at the University of Milan, Italy. 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. In October 2013 she 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. In October 2015 she joined the Research Department at Sidra Medicine. She is the Editor of the section of Translational Cancer Biology of JTM and of the Section Immune Response of Advances in Cancer Biology-Metastasis (Elsevier).



### **Dr. Nicholas van Panhuys**

**Laboratory of  
Immunoregulation**

Dr. van Panhuys completed his BSc in Biochemistry and Molecular Biology, Cell and Developmental Biology at Victoria University (New Zealand). Following this, 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. He was then awarded the NZ Foundation for Research Science and Technology post-doctoral fellowship award, to work as a visiting fellow at the National Institutes of Health (Bethesda, USA) in the Laboratory of Immunology with Dr. Ronald Germain. Consequently he was appointed as a research fellow in the Laboratory of Systems Biology at the NIAD, NIH. Since 2015, he has led the Laboratory of Immunoregulation at Sidra Medicine.



### **Dr. Luis R Saraiva**

**Laboratory of Neurometabolism  
and Functional Genomics**

Dr. Saraiva completed a Licenciatura (BSc+MSc) 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 (summa cum laude). 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 the research branch of Sidra Medicine. Additionally, he is an Adjunct Faculty Member at the Monell Chemical Senses Center (Philadelphia, USA) and at Hamad bin Khalifa University (Doha, Qatar).



### **Dr. Mohammad Haris**

**Laboratory of Molecular  
and Metabolic Imaging**

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 in 2007. Afterwards, he did his postdoctoral fellowship in the department of Radiology at Perelman School of Medicine at the University of Pennsylvania, Philadelphia, USA, and subsequently he became research scientist in the same department. During his tenure at Penn, 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 the research department at Sidra Medicine as a Principal Investigator in October 2013. Also, he holds an Adjunct Professor appointment at Qatar University.



### **Dr. Nico Marr**

**Laboratory of Immunogenetics  
in Infectious**

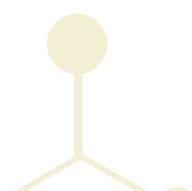
Dr. Nico Marr is a principal investigator leading the Laboratory of Immunogenetics in Infectious Diseases and holds an adjunct faculty appointment with the College of Health and Life Sciences at Hamad bin Khalifa University. He received his doctoral degree from the Julius-Maximilian-University of Würzburg, Germany, for studies in the field of microbiology and immunology. His dissertation and early postdoctoral research at the University of British Columbia in Vancouver, Canada, focused on host-pathogen interactions and immune evasion mechanisms of *Bordetella pertussis*, the etiological agent of whooping cough. Dr. Marr then completed a second postdoctoral fellowship in respiratory health and worked as a research associate at the British Columbia Children's Hospital Research Institute in Vancouver and the Canadian Center for Vaccinology located at the IWK Health Centre in Halifax, investigating human immune defenses in early life against common viral infections. He joined Sidra Medicine as an independent investigator in 2015.



### **Dr. Sara Deola**

**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). 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). Her clinical training was at the BMT Unit of Hospital San Raffaele, and she worked 5 years as staff physician in the Hematology and BMT Department of the General Hospital of Bolzano, Italy, mostly following allogeneic HSCT patients. She joined Sidra Medicine in 2014, as Bone Marrow Transplant Program Manager in Translational Medicine/Research, and she is holding a PI position since June 2017 in the Advanced Cell Therapy Core.





**Dr. Wouter Hendrickx**  
**Cancer Omics**  
**Laboratory**

Dr. Wouter Hendrickx is an investigator in immunology and Inflammation in the Human Immunology Division and member of the Cancer Precision Medicine Working Group at Sidra Medicine. He is the PI of the Pediatric Cancer Omics Laboratory 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). At Sidra he has focused since 2014 on the tumor immune micro environment deploying bio-informatic tools to analyze gene-expression data from bulk tumor for immune related signatures and other determinants of the immune phenotype and translating the findings to the wet lab environment. He was a participant of the EU FP6 and PF7 grant framework and is a 2015 QNRF JSREP awardee. Since 2019 he leads Sidra Medicine's efforts in establishing a biorepository for pediatric cancer patients.



**Dr. Younes Mokrab**  
**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 PhD in Bioinformatics from Prof. Tom Blundell lab, University of Cambridge, UK (2007), followed by a postdoctoral fellowship from Prof. Mark Sansom Lab, University of Oxford (2010). Next, he moved to the Pharmaceutical industry, initially Lonza Biologics performing in silico bio-pharmaceuticals engineering and subsequently Eli Lilly. Upon joining Sidra, Dr. Mokrab helped establish research programs in population and medical genetics and is a co-founding member of the Qatar Genome Program Research Consortium. Also, he co-chaired international meetings including Sidra Functional Genomics and CUDOS. He currently holds academic appointments at WCM-Q and HBKU.



## BREAKOUT STORY

# Clinicians At Sidra Medicine Challenging Conventional Cancer Staging Techniques



When the COVID-19 pandemic halted Sidra Medicine's access to resources that are traditionally used to determine the cancer stage of some of its patients, clinicians at the hospital decisively switched to a more accessible but unconventional technique. Not only was the alternative successful, a recently published study on it led by **Dr. Ata Ur Rehman Maaz**, Senior Attending Physician at Sidra Medicine, and **Dr. Mehdi Djekidel**, Division Lead of Nuclear Medicine and Molecular Imaging at Sidra Medicine, also shows the benefits of utilizing it as the primary means of cancer staging.

Traditionally, an MIBG scan is used for cancer staging of patients with Neuroblastoma, a solid tumor of the nervous system which particularly arises in the neural crest cells of children and is the second most common pediatric cancer in the world. The radiopharmaceutical for an MIBG scan, however, is sourced from large commercial suppliers in Europe. This posed a challenge at the height of the COVID-19 pandemic in Qatar during 2020 when restrictions on flights also halted the supply of the reagent.

When three Neuroblastoma patients at Sidra Medicine needed to be staged, Dr. Maaz and Dr. Djekidel obtained a radiotracer called Gallium DOTATATE from Hamad Medical Corporation to swiftly meet the clinical need of these patients. Gallium DOTATATE scans are mostly used to stage neuroendocrine tumors and sparse reports exist of its effectiveness in Neuroblastoma staging, mostly in the event of cancer relapse.

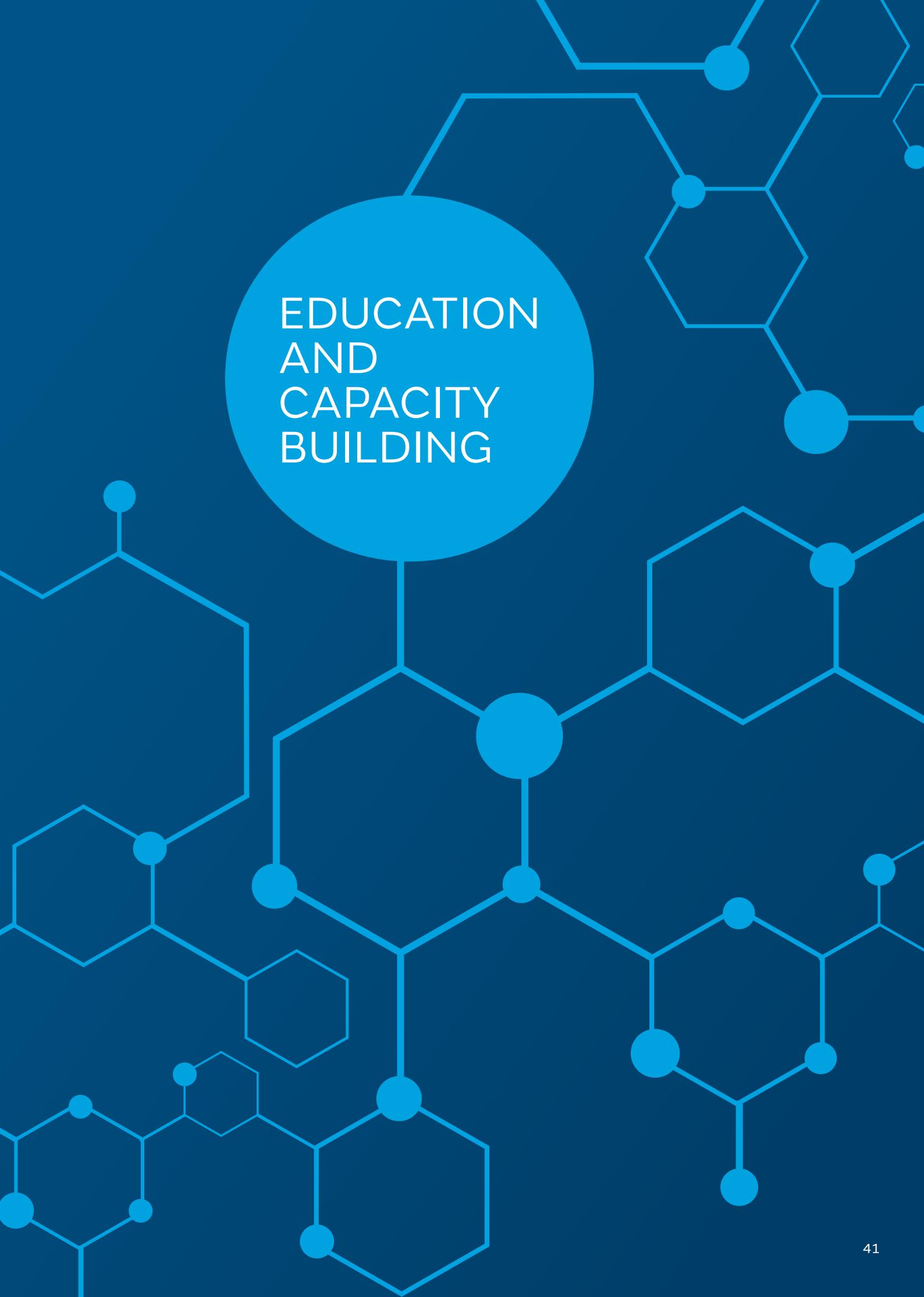
"Sidra Medicine is a forward-looking organization," said Dr. Maaz, "If there is a situation like a COVID-19 lockdown and a patient comes, we are not willing to cut corners and are thinking of cutting-edge technologies to obtain and use."

Although MIBG scans have been the gold standard of cancer staging for the past 40 years, Dr. Maaz and Dr. Djekidel are questioning whether this is the only way of initial cancer staging and whether Gallium DOTATATE can be a viable replacement.

Gallium DOTATATE scans are promising not only for cancer staging, but also cancer therapy in Precision Medicine. First and foremost, its detailed results can give a clearer picture of what level of individualized and targeted care the patient needs, according to Dr. Maaz. On another level, it has potential in Theragnostics, a field of nuclear medicine that pairs diagnostic and therapeutic elements, said Dr. Djekidel.

Dr. Djekidel is collaborating with Dr. Othmane Bouhali, Director of Research Computing at Texas A&M University in Qatar, and Dr. Younes Mokrab, Co-lead of Population Genomics Research Program at Sidra Medicine, to explore advanced imaging techniques such as radiomics and artificial intelligence applications in PET that can further enhance the added value of scans such as Gallium DOTATATE.

**Written by Areesha Lodhi**



EDUCATION  
AND  
CAPACITY  
BUILDING

# CAPACITY BUILDING AT SIDRA RESEARCH

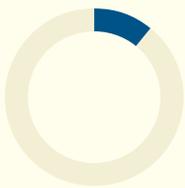
Outreach and education at Sidra Research is paramount, the department hosts several trainees, volunteers, and visiting researchers/scientists coming from various universities and institutions, both locally and from around the world.

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.



52

externs and volunteers enrolled in 2021



6

are PhD students from HBKU



27

are students from Local Universities; including Hamad Bin Khalifa University, Qatar University QU and Northwestern University in Qatar

## 6 VISITING SCIENTISTS:

**Dr. Ahmad Al Ater – Dr. Bernice Lo  
Dr. Ana Cláudia – Dr. Luis Saraiva  
Dr. Alice Turdo – Dr. Cristina Macalli  
Dr. Marcia Baldoni – Dr. Sara Tomei  
Dr. Farooq Al Ajli – Dr. Khalid Fakhro  
Dr. Khadega Adel – Dr. Chiara Cugno**



# LEADERS IN SCIENCE

## SIDRA RESEARCH SERIES

Science is a global endeavour to help speed up research projects and give life to new ideas and scientific breakthroughs. Educational talks are an integral part helping researchers 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. Sidra Research Series is a platform that helps boost collaboration and bridge the gap between research and practice.



The series this year hosted a multitude of local and international speaker:

#### INTERNATIONAL SPEAKERS:

##### **Dr. Junaid Kashir**

Assistant Professor in Clinical Embryology  
*Alfaisal University, KSA*

##### **Dr. Elvira Verduci**

Pediatrician  
*University of Milan, Italy*

##### **Dr. Jiwon Sarah Choi**

Chief of Technology  
*Div. of R&D G+FLAS Life Sciences, Korea*

##### **Dr. Fabeha Fazal**

Associate Professor in Pediatrics  
*University of Rochester, USA*

##### **Dr. Manoj Kumar**

Department of Neuroimaging  
and Interventional Radiology (NIIR)  
*National Institute of Mental Health and  
Neurosciences (NIMHANS), Bangalore, INDIA*

##### **Dr. Matteo Avella**

Assistant Professor of Genetics  
*University of Tulsa, USA*

##### **Dr. Saquib A Lakhani**

Associate Professor  
*Yale University School of Medicine, USA*

##### **Dr. Saurabh Jha**

Associate Professor  
*University of Pennsylvania, USA*

**LOCAL SPEAKERS INSTITUTIONS ACROSS QATAR:**

**Dr. Julia Reichelt**

Senior Academic Research Scientist and Principal Investigator at Translational Research Institute Hamad Medical Corporation

**Dr. Essam Abdelalim**

Assistant Professor  
HBKU

**Dr. Khalid Hussein**

Division Chief of Endocrinology  
Sidra Medicine

**Dr. Sawssan Ahmed**

Psychologist  
Sidra Medicine

**Dr. Donald Love**

Division Chief-Pathology Genetics  
Sidra Medicine

**STAFF SCIENTISTS:**

**Dr. Andrea Guennon**

**Dr. Alexandra Marr**

**Dr. Mathieu Garand**

**Dr. Shana Jacob**

**Dr. Meritxell Espino Guarch**

**Dr. Rafah Mackeh**

**Dr. Christophe Raynaud**



**59 Talks  
for 2021**

**PIS:**

**Dr. Nico Marr**

**Dr. Nicholas Van Panhuys**

**Dr. Wouter R. Hendrickx**

**Dr. Bernice Lo**

**Dr. Luis Miguel Rodrigues Saraiva**

**Dr. Sara Tomei**

**Dr. Stephan Lorenz**

# Student Testimonials



## **Asma Saeed**

**Extern with Ammira Akil**

My passion for knowledge lead me to seek an externship opportunity in the advanced research department of Sidra Medicine. Having completed my undergrad education from Monash University Australia in Malaysia, I was intrigued by Sidra Medicine's capability of incorporating 'Precision Medicine' in patient care.

I joined Dr. Ammira Akil's 'Precision Medicine in Diabetes Prevention' program where I was able to learn and gain valuable hands-on experience on the genomic techniques involved. As a fresh graduate, this hands-on training to develop skills is extremely important. I was involved in wet lab procedures and given equal opportunity for dry lab research training. It is a homogenous pool of opportunities for young entry-level researchers.

I ended up with an even greater opportunity of being able to work on a unique project and learn something new and interesting in the dry lab research area. I will carry on this knowledge with me and be more open to participate in more dry lab projects and not restrict myself to only one research sector.

I would recommend and even encourage other fresh graduates to also participate in Sidra Medicine's externship program. It is an incredible opportunity for learning from highly skilled senior researchers, gaining hands-on experience, mastering new unique techniques, as well as, an early opening for young researchers to be part of the scientific community.



## **Amira Kohil**

**Extern with Annalisa Terranegra**

I was given the opportunity to join the externship program in Sidra Medicine through my QU supervisors while studying my Master's in Science degree. I was particularly interested in Dr. Annalisa Terranegra's research as it focused on nutrigenomics and gut microbiome impact on different disorders. In these two years I was at Sidra as a research trainee to complete my thesis' research, I gained a lot of experience in different experimental and computational procedures, mainly mentored by Dr. Arun Lakshmanan. The members of the nutrigenomics team were always available and supportive and provided me with all the support to complete my thesis and enrich my research experience. I was also given the opportunity to present my project in different local and international conferences. Training at Sidra enhanced my academic writing, leading to publications as first author and co-author. This all enhanced my experience in the different aspects of research. Finally, my advice for future trainees is to try to benefit the best from their time at Sidra in terms of scientific work and good friendship.



**Areesha Lodhi**  
Extern with Noor Faisal

As an aspiring science and health reporter, my externship at Sidra Medicine provided me with an in-depth opportunity to excel in the field and develop interpersonal skills. I worked in a challenging setting all while having supportive supervisors who trusted my abilities provided guidance.

The structure of the externship facilitated my learning, sense of discipline, and the ability to reflect on my work and improve it.

With the support of abundant sources facilitating my understanding of concepts and supervisors who provided thorough feedback, I was able judge on how to present information depending on who the target reader is, thereby ensuring accessible and engaging content. The most interesting part was speaking to the brilliant scientists and researchers at Sidra Medicine who are inspirationally passionate about sharing the innovative work they do.



**Eman Wehedy Abdelsalam**  
PhD student with Souhaila Al Khodor

I am pleased to do my Ph.D. project in Sidra Medicine; all the facilities are especially well-equipped and helped me achieve my work. The place is so comfortable, and the intelligent scientists are approachable and happy to help. I was working at Qatar university on a project which is a continuation of my area of interest using advanced approaches.

My Principal Investigator Dr. Souhaila Al Khodor is very cooperative and organized; she helped me achieve my work. The team is very friendly and ready to assist at any time. The experience I gained during my externship period will help me have a good position in my future career.

I highly recommend junior scientists and graduate students to join Sidra medicine to do their research.



**Muhammad Kohailan**  
PhD student with Khalid Fakhro

It started in Sep 2018, when I joined the PhD program in “Biological and Biomedical Sciences” at Hamad Bin Khalifa University. Seeking a mentor to supervise my academic journey, attending the World Innovation Summit for Health (WISH) in Nov 2018 allowed me to discover the amazing work being performed by Dr. Khalid Fakhro and his team. Immediately, on the same day of the conference, I contacted Dr. Khalid, requesting to explore the lab and the several projects they work on.

With a background in Biochemistry and basic science, I decided to shift gears and explore the world of bioinformatics after being introduced to the “big data” analysis carried out by Dr. Khalid’s team and others at Sidra Research. The cooperative, professional and friendly working environment Helped make my decision clear.

My PhD project focuses on detecting human germline mutations found in children but not in their parents, so called de novo mutations, which in term are responsible for a high number of severe diseases.

Being in one of the fast-developing organizations in Qatar and the world, my prospective future career is enhanced and expanded with more opportunities in my scientific road. I believe Sidra Medicine is the right place for junior students who wish to gain an excellent multidisciplinary experience in research.



**Shaikha Al Sayegh**  
MSc student with Ammira Akil

I have always heard a lot about Sidra Medicine Research Branch and was always looking forward for a chance to be part of this great institution. After completing my master’s degree I immediately applied to join Sidra Research for an opportunity to improve my research skills and knowledge in the medical research field.

I was involved in two projects while training in Dr. Ammira’s lab, which was really exciting, as was meeting new people who are talented and experienced scientists inspired me to be ambitious in everything. The highlight of my experience is that I got to deal with DNA and RNA extraction and protein extractions, as well as, real time-PCR. I gained valuable knowledge in this period of time, and felt a powerful urge to learn more. Sidra affords you the best equipment and devices in Qatar for working on projects. I feel lucky have been trained by Dr. Ammira, she’s one of the most intelligent people I have ever met in science fields, very supportive as well. All the respect to her and to all Sidra staff.



**Fajr Almarzooqi  
and Sara Almarzooqi**  
**Graduate Associates for the year 2021**

Within a few weeks of joining Sidra Medicine Research Branch as Graduate Associates, we managed to take an active role in ongoing projects under the supervision of Dr. Souhaila Alkhodor. We were encouraged to participate in scientific forums such as Research Day and PMFG. Initially, we were hesitant as the experience was completely new, but we pushed ourselves out of our comfort zone and decided to seize the amazing opportunities offered to us. Research is so much more than what is done in the labs. We participated by attending, gaining knowledge, being aware of what research is recently being done at Sidra and around the world. It was a proud moment to present a poster on the work we achieved in such a short period of time. At Sidra, and with our experience so far, we know that the best is yet to come. Our experience would not have been the same without such a supportive PI and colleagues. Our exposure has been greatly complemented by prior experience in the lab, making the transition so much smoother. Our advice for future graduate associates is to have enough lab experience during their undergraduate studies, to ask questions, and to always challenge yourself to jump out of your comfort zone.

## BREAKOUT STORY

# Loss of Smell as the Best Predictor of COVID-19



Sidra Medicine is one of the leading academic medical centers participating in the global effort to tackle COVID-19-related smell and taste loss. **Dr. Luis R. Saraiva**, Principal Investigator at Sidra Medicine, is a leading member in the Global Consortium for Chemosensory Research (GCCR), and more recently integrated into its Leadership Team. The GCCR conducts worldwide scientific studies to assess possible relationships between respiratory illnesses (e.g., COVID-19, influenza or the common cold) and their effects on smell and taste.

“One of our goals was to see if we could help predict the onset of COVID-19 by associating loss of smell with the onset of infection. The results would bring awareness to people that if they are suffering from this set of symptoms, they should self-isolate to avoid spreading the virus, and seek professional medical testing and treatment immediately,” said Dr. Saraiva.

His study collected responses from all over the world via online surveys that have been translated into 35 languages, including Arabic. Most published COVID-related research studies are conducted in Europe and North America which can neglect other regions such as the Gulf region. The inclusion of Arabic-speaking participants creates diversity and increased reliability.

Qatar is one of the leading countries investing heavily in COVID-19-related research projects in collaboration with national institutions during

the pandemic. With the recent appointment of Dr. Saraiva to the Leadership Team of the GCCR, Sidra Medicine is distinguishing itself from other regional and national institutions by addressing one of the most pressing health crises worldwide.

“The MENA region and its people are massively underrepresented in scientific studies. To prevent that from happening in the GCCR studies, we ensured that the survey was made available in Arabic, so that it can be disseminated in Qatar and the broader MENA region. Ultimately, this research contributes to the prevention of COVID-19 not only in Qatar or the MENA region, but also worldwide. We live in an age where air travel is more accessible than ever before—we are all connected.” added Dr. Saraiva.

This research revealed the ODoR-19 test as an important self-screening tool, which the medical community could implement in a clinical setting, and the general population can use at home to help prevent the spread of COVID-19.

Dr. Saraiva’s contribution to the research looks at the COVID-19 pandemic from a global perspective as the symptoms are linked with individual genetic backgrounds as well as environmental factors. As the leading Qatari institution in Precision Medicine, Sidra Medicine hopes to collaborate with other global organizations such as GCCR to discover more about COVID-19.

**Written by Jiwon Seo**

## BREAKOUT STORY

# Validation of a Pathogenic Variant of Bruton's Tyrosine Kinase (BTK) gene in X-Linked Agammaglobulinemia (XLA)



As one of the leading academic medical centers in Qatar, Sidra Medicine promotes tight integration between clinicians and researchers in ongoing Precision Medicine-related studies. This research was no exception. **Dr. Mehdi Adeli**, Division Chief of Pediatric Allergy and Immunology astutely identified a patient with a BTK variant of uncertain significance and collaborated with **Dr. Bernice Lo** and her team of researchers for further investigation to determine functional impact of the variant.

"Sidra Medicine provides the optimal environment for Precision Medicine by combining clinical teams that are experts in looking at the phenotype and researchers who can conduct the necessary experiments. Sidra Medicine is supportive to clinicians and researchers in working together to find the best way to help the children," said Dr. Adeli.

X-linked agammaglobulinemia, also known as XLA is a rare inherited immunodeficiency disease caused by BTK gene mutations compromising the person's natural ability to fight off infections. While it is mostly males who are affected, the disease can also affect a female minority.

Children with XLA are usually healthy for the first 1 or 2 months as their bodies are protected by antibodies acquired before birth from their mother. A child with XLA cannot make antibodies to defend against bacterial and viral infections. Most XLA-related infections occur in the middle ear, sinuses, and lungs.

A fully Sidra Medicine-funded research study involving Dr. Adeli along with Dr. Lo and her team confirmed the link between a specific variant of the BTK gene and XLA, resulting in a recently published article, "X-Linked Agammaglobulinemia Case with TH Domain Missense Mutation in Bruton Tyrosine Kinase."

Dr. Adeli's patient was a three-year-old male patient with a history of pseudomonas (a type of germ) infections at 1 year of age and had a mutation in the BTK gene. The gene was passed down from his mother, indicating that she was a carrier. The variant found in the patient was originally categorized as a "variant of uncertain significance" which meant that genetic data alone was insufficient to declare the variant as pathogenic. The researchers decided to conduct flow cytometry staining to determine BTK protein expression. The result showed that the patient had a BTK deficiency and that the mother had some cells that exhibited normal BTK expression and some cells with deficient BTK expression, similar to her son. Together, these findings provided functional evidence that the variant was pathogenic.

Functional evidence is essential to determine the pathogenicity of a variant of uncertain significance. Clarifying the pathogenicity of the variant will help future pediatric patients who carry the same variant get prompt and proper treatment.

"Having a precise or definitive diagnosis means that the patient can be treated immediately with the proper therapy for this particular disease and prevent the patient from developing any worse complications," said Dr. Lo.

Primary immunodeficiency diseases (PID) are not rare in children in Qatar as there is an estimated prevalence of 4.7 PID patients per 100,000 children younger than 14 years of age whereas it is estimated to be 1.1 per 100,000 children younger than 19 in Asia. A scientific study published in 2013 outlines that 23.7% of 131 patients had antibody deficiency, making it the most common type of PID in Qatar. Hence, Sidra Medicine is one of the few national institutions at the forefront in raising awareness of these diseases.

## BREAKOUT STORY

# Sidra Medicine Clinicians Conduct Breakthrough Research with In-house Resources



From being the first to study the molecular epidemiology of a group of highly resistant bacteria in the Gulf region to examining the impact of school closures during COVID-19 on the spread of influenza, clinicians and researchers at Sidra Medicine are collaborating to make breakthroughs in research on public health issues while solely using the resources made available by the institution. New diagnostic techniques studied identify an infection within an hour, compared to the two to three days taken by conventional methods, and also indicate whether the pathogen is resistant to an antibiotic or not.

“Sidra Medicine’s vision is to be a beacon of knowledge,” said **Dr. Andres Lopez**, senior attending physician in Microbiology. “We are here not only for Qatar but also for the entire region.” Clinicians have received grants from Sidra Medicine’s Internal Research Fund and utilized solely on-site resources, staff, and patients.

In 2018, Sidra Medicine awarded an Internal Research Fund grant to Dr. Lopez to study extended-spectrum beta-lactamases, enzymes found in some bacteria strains that make them harder to treat. In Qatar and the Arabian Gulf region, these bacteria’s resistance mechanisms pose a significant threat to the population and cannot be killed with commonly prescribed antibiotics. Although the study focused on Qatar’s pediatric population, its results could be applied to people in other GCC countries because of similar demographics and the high prevalence of the infection here, according to Dr. Lopez.

Following the outbreak of COVID-19, another Internal Research Fund grant enabled Dr. Lopez to study how school closures meant to curb the virus would

also reduce the transmission of influenza A. Dr. Lopez and his team were the first to produce such a report globally. Moreover, this phenomenon was later documented by other countries during the pandemic.

Diagnosis mechanisms developed in Dr. Lopez’s studies emphasize the use of Precision Medicine to tailor to patients’ specific genetic makeup. Research department facilities have especially helped with this process. In the case of life-threatening bloodstream infections like sepsis, the rapid diagnostic techniques being studied and implemented could save a patient’s life through Precision Medicine by prescribing the best possible antibiotic in time.

Alongside the internal funding and resources, Sidra Medicine’s patient pool is large and varied, leading to reliable results. “The diversity of the ex-pat population in Qatar allows us to embrace and to study several possible variants,” said Dr. Lopez.

The participation of Sidra Medicine patients enables clinicians to understand their characteristics better and, therefore, enhance the patient experience here. “This [research] gives you better information about your patient population and that is important for their management,” said Dr. Mohammad Rubayet Hasan, clinical molecular microbiologist at Sidra and a co-author on Dr. Lopez’s papers.

Dr. Lopez is continuing to work with other clinicians and researchers with 100% in-house resources to conduct leading research on the region’s public health, especially for women and children.

**Written by Areesha Lodhi**





# GRANTS AND COLLABORATIONS

# GRANTS AND COLLABORATIONS

12 total grants from Qatar National Research Fund (QNRF) and other external funds were awarded in the last 18 months

**Totaling QR 7.3 M**

## BREAKDOWN OF GRANT APPLICATIONS IN THE LAST 18 MONTHS

2020 Grants Applications	Submitted	Awarded
National Priorities Research Program (NPRP-13S)	24	6
National Priorities Research Program (NPRP-13C)	2	0
Path towards Precision Medicine Call (PPM04)	12	2

2021 Grants Applications	Submitted	Awarded
National Priorities Research Program (NPRP-14S)	7	tba
NPRP – Blue Skies Research Award (BSRA)	3	tba
Path towards Precision Medicine Call (PPM05)	8	tba
Equine Research Call	1	tba
Rapid Response Call (RRC) Covid-19 cycle 2	1	0
Graduate Sponsorship Research Award (GSRA8)	4	1
Graduate Sponsorship Research Award (GSRA8+)	2	1
Postdoctoral Research Award (PDRA7)	3	2
Early Career Researcher Award (ECRA3)	1	0
High School Research Experience Program (HSREP3)	1	1
Conference and Workshop Sponsorship Program (CWSP20)	1	1
Conference and Workshop Sponsorship Program (CWSP21)	2	tba
Researchers Exchange and Mobility Program (REMP3)	2	1
Global Grants for Gut Health	1	tba
CONquer canCER Now Grants	1	tba
Gilead Research Scholars in Liver Disease	1	tba
Phizer Quality Improvement grants	1	tba
Reimagine Biomedical Research for a Healthier Future	1	tba
SOLVE-RD – Solving the Unsolved Rare Diseases	1	1
Amryt IIS Award	1	1



**Asma Al Sulaiti**, Research Specialist in the Laboratory of Immune Biological Therapy recently completed her master's degree from Qatar University in MSC in Biological Medical Sciences.

**Reem Ahmadmojahid Hasnah**, research specialist in the Laboratory of Neurometabolism and Functional Genomics recently completed her master's degree from Hamad bin Khalifa University in MS in Biological and Biomedical Sciences.



**Alya Alawi Al-Kurbi**, Research Specialist in the Laboratory of Genomic Medicine recently completed her master's degree from Hamad bin Khalifa University in MS in Biological and Biomedical Sciences.

**Khalid Jabir Al-Lakhen** from Laboratory of Immunoregulation recently completed his master's degree from University at Albany in Molecular, Cellular, Developmental and Neural Biology (MCDN).



# Top Ten Original Publications

**1** *Humans with inherited T cell CD28 deficiency are susceptible to skin papillomaviruses but are otherwise healthy*

**AUTHORS**

Tanwir Habib,  
Andrea Guennoun,  
Taushif Khan, Mahbuba  
Rahman, Fatima Al Ali,  
Manar Ata, Nico Marr

**JOURNAL**  
CELL

**2** *Inherited PD-1 deficiency underlies tuberculosis and autoimmunity in a child*

**AUTHORS**

Taushif Khan,  
Fatima Al Ali,  
Mahbuba Rahman,  
Nico Marr

**JOURNAL**  
NATURE MEDICINE

**3** *Fasting-mimicking diet is safe and reshapes metabolism and antitumor immunity in cancer patients*

**AUTHORS**

Darawan Rinchai,  
Davide Bedognetti

**JOURNAL**  
CANCER DISCOVERY

**4** *Germline genetic contribution to the immune landscape of cancer*

**AUTHORS**

Wouter Hendrickx,  
Jessica Roelands,  
Younes Mokrab,  
Najeeb Syed,  
Davide Bedognetti

**JOURNAL**  
IMMUNITY

**5** *X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19*

**AUTHORS**

Nico Marr, Manar Ata,  
Fatima Al Ali,  
Taushif Khan

**JOURNAL**  
SCIENCE  
IMMUNOLOGY

**6** *2D MXenes with antiviral and immunomodulatory properties: a pilot study against SARS-CoV-2*

**AUTHORS**

Laura Fusco,  
Jean-Charles Grivel,  
Davide Bedognetti

**JOURNAL**  
NANO TODAY

**7** *Biallelic variants in SLC38A3 encoding a glutamine transporter cause epileptic encephalopathy*

**AUTHORS**

Aljazi Al-Maraghi,  
Khalid A Fakhro

**JOURNAL**  
BRAIN

**8** *Development of a fixed module repertoire for the analysis and interpretation of blood transcriptome data*

**AUTHORS**

Darawan Rinchai,  
Mohammed Toufiq,  
Mathieu Garand,  
Basirudeen Syed  
Ahamed Kabeer,  
Mohamed Alfaki,  
Davide Bedognetti,  
Damien Chaussabel

**JOURNAL**  
NATURE  
COMMUNICATIONS

**9** *Thousands of Qatari genomes inform human migration history and improve imputation of Arab haplotypes*

**AUTHORS**

QGP Research  
Consortium,  
Elbay Aliyev, Sara Tomei,  
Stephan Lorenz,  
Wei Liu, Najeeb Syed,  
Ramzi Temanni,  
Tariq Abu Saqri,  
Tariq Abu Zaid,  
Tushar Pathare,  
Shafeeq Poolat,  
Rashid Al-Ali,  
Souhaila Al-Khodor,  
Xavier Estivill,  
Khalid A Fakhro,  
Younes Mokrab,  
Jithesh V Puthen,  
Zohreh Tatari

**JOURNAL**  
NATURE  
COMMUNICATIONS

**10** *Inherited human c-Rel deficiency disrupts myeloid and lymphoid immunity to multiple infectious agents*

**AUTHORS**

Tanwir Habib,  
Luis R Saraiva,  
Eman A Moussa,  
Andrea Guennoun,  
Nico Marr

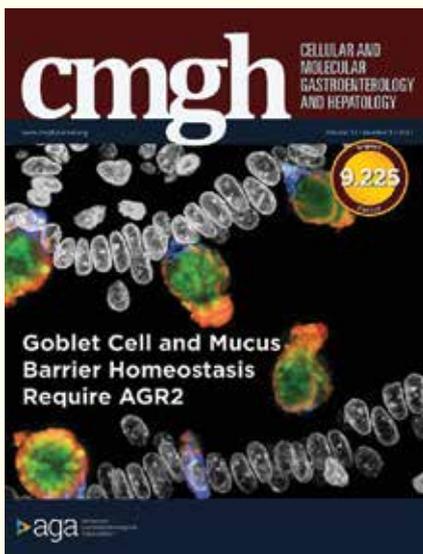
**JOURNAL**  
JOURNAL  
OF CLINICAL  
INVESTIGATION



# Sidra Medicine Publications on Journal Covers

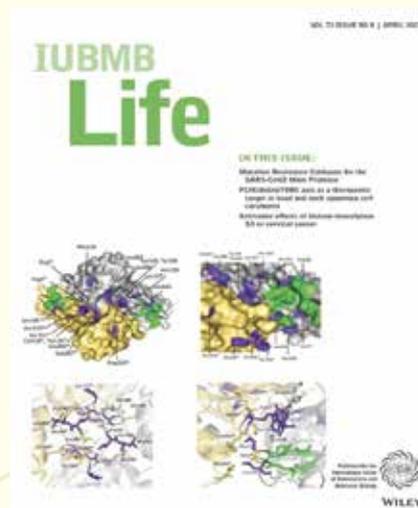
## Dr. Bernice Lo

Dr. Bernice and her team on the journal cover of Cellular and Molecular Gastroenterology and Hepatology for their study on *Human AGR2 Deficiency Causes Mucus Barrier Dysfunction and Infantile Inflammatory Bowel Disease*.



## Dr. Navaneethakrishnan Krishnamoorthy

Dr. Navaneethakrishnan Krishnamoorthy and Dr. Khalid Fakhro on the journal cover of IUBMB Life for identifying mutation “coldspots” in the #SARSCoV2 main protease.



## Dr. Nico Marr

Dr. Nico Marr and his team on the cover of JCI Insight for a comprehensive analysis of antibody responses in children and adults to the four endemic human coronaviruses.



## BREAKOUT STORY

# Qatar's First Diabetes Mellitus Database



There are 1.1 million children and adolescents living with type 1 diabetes, according to the International Diabetes Federation Diabetes Atlas 9th edition.

Diabetes mellitus is a disorder in which the body does not produce enough or respond normally to insulin, causing blood sugar (glucose) levels to rise and is classified into different types based on pathogenesis. The different types include type 1 diabetes, type 2 diabetes, neonatal diabetes mellitus (NDM), maturity-onset diabetes of the young (MODY), syndromic forms of diabetes mellitus and some yet unclassified forms of diabetes mellitus.

The Middle East and North Africa (MENA region) has one of the highest cases of childhood diabetes mellitus in the world. The number of children with type 1 diabetes is estimated to be 60,700 with 10,200 newly diagnosed each year.

Much of diabetes studies in the MENA region, including Qatar, are only retrospective reports, case studies with very few prospective studies and some questionnaire-based studies and not enough data on epidemiology, genetic and molecular mechanisms of the disorder. Qatar considers diabetes mellitus as one of its major health concerns due to changes in lifestyle and dietary habits.

To collect data on epidemiology, genetic and molecular mechanisms on children and adolescent patients with diabetes mellitus, **Dr. Khalid Hussain's** clinical research team at Sidra Medicine successfully compiled and analyzed data in a report funded by Qatar National Research Fund: "Epidemiology, genetic landscape and

classification of childhood diabetes mellitus in the State of Qatar."

Every diabetes mellitus patient aged 0–18 years was recruited from 2018–2020. Sidra Medicine is the only childhood diabetes center in Qatar, hence all children with diabetes mellitus were referred there, allowing to capture all children diagnosed with diabetes mellitus in the present study.

The results from the study show that there are 1,325 children diagnosed with diabetes mellitus in Qatar. Among the pediatric patients, type 1 diabetes was the most common diabetes among children with 83%. As for the ethnicity among children with diabetes mellitus, Qatari population recorded the highest incidence with 51.59%.

The collected data is a major contribution to Precision Medicine as clinicians and researchers have accurate data on people with diabetes in Qatar to allow implementation of present and future therapies for all different types of childhood diabetes. As Sidra Medicine is an academic medical center, collaboration between clinicians and researchers opens doors to further studies that help develop strategies for managing different types of diabetes.

Every medical care is given to patients with the goal of complete recovery. However, clinicians at Sidra Medicine also ensure that quality of life of patients is not impaired while being treated. Ongoing studies that tailor to patients' needs allow continued improvements in present and future patient care.

**Written by Jiwon Seo**



# EVENTS/ HIGHLIGHTS

# EVENTS/HIGHLIGHTS

## Al Jazeera Arabic’s “Book of Life,” about Sidra Medicine and our QF partner’s role in advancing Qatar’s Precision Medicine Program

What if we are able to read the human body with more accuracy? What if we are able to prepare ourselves to withstand new epidemics? What if we are able to tailor-make healthcare treatments that will prevent our children, our parents, our families and friends from running pillar to post looking for answers to their health problems?

In an attempt to answer these questions, Qatar Foundation (QF) teamed up with Al Jazeera to produce a documentary on precision medicine titled *The Book of Life—Precision Medicine*, which showcases QF’s efforts and investments in this area, which is said to be the future of healthcare.

Precision medicine is an approach for disease treatment that factors in individual differences in genes, environment, and lifestyle for each person. Why do some people react differently to certain drugs, or why are some people becoming severely ill with SARS-CoV-2 whereas others are showing no symptoms—these are questions precision medicine seeks to answer, thereby offering accurate diagnosis and effective treatment.

“Precision medicine is to provide medical care to a person based on their genetic make-up. Different treatment options are to be provided—whether diagnostic, therapeutic, or preventive—based on the individual person’s genetic composition. The time of ‘one size fits all’ will become history,” Dr. Said Ismail, Director of Qatar Genome Programme (QGP), a member of Qatar Foundation Research, Development and Innovation, said.

Despite significant scientific advances in the field of medicine, unknown problematic areas continue exist and baffle doctors. One such example is that of baby Shereen. At only five months, she began to fall ill with severe chest infections. She was given a host of medicines, including antibiotics, and put on oxygen and even given sedatives, but her little body was just not responding to any of those treatments. Her condition kept deteriorating until she reached a stage where she couldn’t breathe on her own and had to be put on a ventilator.

As the baby kept fighting for her life, she was referred to QF’s Sidra Medicine. Here, doctors ran some tests, including some genetic tests, and it was discovered that the baby had a defect in a specific gene which affected her immunity. This was enough for the doctors to give her the precise treatment for her condition.

Baby Shereen has been on a road to recovery since.

Genomics technology is one of the most prominent techniques that help achieve the mission of precision medicine, according to Dr. Khalid Fakhro, Acting Chief Research Officer at Sidra Medicine and the Director of the Precision Medicine program at the healthcare center. “What distinguishes precision medicine from traditional medicine is the availability of appropriate technologies to diagnose the disease at a molecular level.”

Precision medicine not only contributes to the treatment of diseases, it has also played a vital role in exploring the severity of the COVID-19 pandemic, and to help reduce its spread.

Dr. Hamdi Mbarek, Scientific and Industry Partnerships Manager, QGP, said: “COVID-19 is now of paramount importance in our scientific research at QGP, where we are supporting State efforts in reducing the spread of this virus by conducting research and developing innovations to understand the human genome in explaining the risks and susceptibility of being infected with COVID-19.”

Genomics and its applications in the field of precision medicine is making great strides in Qatar. Precision medicine has been identified as one of the national priorities—an aspect that will be critical in shaping the future of humanity.



## Sidra Medicine leads research study, in collaboration with HBKU's Qatar Computing Research Institute & University of California San Francisco, representing a significant step toward personalized cancer immunotherapeutic approaches

Sidra Medicine and Qatar Computing Research Institute (QCRI) at Hamad Bin Khalifa University (HBKU), Qatar Foundation, led a research study with the University of California San Francisco (UCSF) that represents a significant step toward personalized cancer immunotherapeutic approaches.

The international team of cancer immunologists, computational scientists, oncologists, biologists, and geneticists found that pre-existing anti-cancer immunity depends heavily on a patient's genetic background. As such, certain genetic variants that make each of us unique can also influence the way the immune system fights tumors. Immunotherapy, a therapeutic approach based on boosting the immune system, has changed the way cancer is treated, yet only a minority of patients respond to the treatment.

The groundbreaking research study, published in *Immunity* (CellPress), one of the top scientific journals worldwide, answers a critical question that has been facing scientists over the past ten years. That is, why some patients develop a spontaneous, yet partial, anti-cancer immunity that makes them more likely to respond to immunotherapy and whether this response is caused by genetic variation in the DNA of the patients.

Dr. Davide Bedognetti, Director of the Cancer Research Department at Sidra Medicine and Adjunct Associate Professor at the College of Health and Life Sciences at HBKU, with Dr. Elad Ziv, Professor of Medicine at UCSF, led the research team as co-senior authors. QCRI's Dr. Mohamad Saad and UCSF's Dr. Rosalyn Sayaman, co-first authors, were the lead computational scientists, with other team members from Sidra Medicine including Dr. Wouter Hendrickx, Jessica Roelands, Dr. Younes Mokrab and Najeeb Syed. The immunogenomic analytic approach used in the study to dissect tumor-host interplay was also implemented as a part of one Qatar National Research Fund's National Priorities Research Program project.

The new joint research holds significant potential for further achievements. Future studies will determine whether a combined "immunogenetic score" can detect patients more likely to benefit from specific immunotherapies, for a truly personalized approach.

Dr. Davide Bedognetti said: "Translating findings into clinical practice to develop personalized immunotherapeutic approaches accounting for patients' genetic fingerprints represents the next challenge. We are now characterizing pediatric cancer patients genetically and immunologically to expand immunotherapy to this population."

Commenting on how the study has highlighted the role of computational analysis in tackling major diseases, Dr. Mohamad Saad said: "We analyzed a set of around 9,000 patients with 30 different cancer types. As the amount and type of data will grow exponentially due to technological advances, machine learning and artificial intelligence methods will be needed to understand them and extract clinically relevant information."

## Research team led by Dr. Souhaila Al Khodor developed the first salivary microbiome composition of the Qatari population. Another great program that supports our personalised preventative medicine program



What our saliva does for us is not always appreciated. As well as helping us digest our food, saliva carries information about the physiological state of the body. This means real-time monitoring of salivary data can provide useful translational clinical applications for detecting various diseases.

The shift toward personalized, or precision, medicine can be propelled by advances in diagnostic tools. One of these is saliva diagnostics. Utilizing saliva to identify and measure biomarkers has the potential to enable highly individualized diagnosis, prognosis and treatment.

“Saliva acts as a mirror for the body’s health,” says Dr. Souhaila Al Khodor, Director of the Maternal and Child Health Program in the Research Department at Qatar Foundation (QF) member Sidra Medicine—a women’s and children’s hospital that has precision medicine built into its core pillars of patient care, research and medical education.

Saliva is composed of RNA, DNA, proteins, electrolytes, metabolites, and microbiota. Variation in either of these components can indicate a change in the health of an individual. The salivary microbiome not only differs from disease to disease, but it also changes based on ethnicity. This is partly due to differences in genetics, diet and environmental factors.

The degree of variation in the salivary biome at a population level has not been studied extensively, and the few studies that do exist do not take into account the Arab population or Qatari population.

Now Dr. Al Khodor’s group has successfully reported the first-ever salivary microbiome composition in the Qatari population, using data from 1,000 individuals participating in the Qatar Genome Programme (QGP). “If data representing Arabs is missing, the first responsibility to fill that gap lies with us as Arab scientists.”

To advance the field of saliva diagnostics, it is vital to create a comprehensive database of biomarkers that signal the presence of a disorder in saliva. Once fully catalogued, changes in biomarker levels can play a key role in maintaining well-being and early detection of diseases.

Bodily fluids like blood and urine have been used in diagnostics for decades, but there are reasons why scientists are now turning to saliva as well. “Saliva is one of the most ideal diagnostic tools,” says Dr. Al Khodor. “It is inexpensive, noninvasive, and easy to handle. More importantly, minimal patient discomfort makes it a favorable choice over other bodily fluids.

Given their speed and cost effectiveness, salivary-based diagnostic techniques can potentially allow screening of an entire population for a specific disease in a timely fashion. Validated salivary biomarkers combined with powerful detection tools have the potential to open a new innovative frontier in personalized healthcare. In the future, salivary tests may pave the way for chair-side diagnosis of multiple diseases, allowing real-time health monitoring, leading to personalized preventative medicine.



## Leading Genomics Prediction Strategies for Health Complications Resulting from Type 2 Diabetes

“Had we detected the disease earlier, we would have been able to prevent such serious consequences.” This is the comment patients have had to hear all too often. At Sidra Medicine, however, researchers and clinicians are helping do just that. They are working together to prevent serious consequences for the most prevalent disease in Qatar—diabetes.

A prediction model called ‘polygenic risk scores’ is becoming popular around the world for assessing the risk of several complex diseases through whole genome sequencing. For the first time, this approach is being used to assess the risk of severe health complications, such as cardiovascular diseases and kidney failure, in Qatari patients with type 2 diabetes, said Dr. Ammira Akil, the Lead Principal Investigator of this research project and the Translational Genomics of Diabetes research team leader at Sidra Medicine. The study is being funded by Qatar National Research Fund and another local partners and is in collaboration with Weill Cornell Medicine in Qatar, Montreal University and Centre of Genomics and Policy at McGill University in Canada.

“Pioneering such studies has great advantage, such as opening the proverbial door for more investigations of this kind,” said Dr. Akil, adding that this will place Sidra Medicine as a leader in genomic prediction of type 2 diabetes complications.

This prediction model is novel because of how it can detect patients at high risk, in whom early interventions would be most beneficial, much earlier than traditional approaches can, according to Dr. Akil.

Due to the polygenic risk scores’ specificity of looking at the risk of any individual, the study will greatly enhance the process of treating diabetic patients through Precision Medicine, particularly Precision Prevention, at Sidra Medicine. “If that risk score identifies individuals who will have a rapid progression of diabetes and its complications, then that enables early intervention to limit the severity of complications,” said Dr. Shahrad Taheri, professor of medicine at Weill Cornell Qatar and a clinical investigator on the study.

The formation of the polygenic risk scores model for this study is made possible by genome sequencing through highperformance computing clusters housed at Sidra Medicine, according to Dr. Ikhlaq Ahmed, the project’s bioinformatician at Sidra Medicine’s research department.

Severe health complications can be the costliest part of diabetes, according to Dr. Taheri. Dialysis due to kidney failure, for example, can cost about \$ 30,000 a year. “If you take that into account with about 20% of the population, so it can be very expensive,” he said.

With a novel approaches and advanced technologies, researchers at Sidra Medicine are tackling severe outcomes of Qatar’s most prevalent disease at their core. This innovative form of Precision Medicine may reduce the burden on hospitals around the country such as Sidra Medicine in the years to come.

**Written by Areesha Lodhi**



## Researchers at Sidra Medicine have identified spots within the novel coronavirus SARS-CoV2 that do not mutate; paving the way for future vaccine design that can address variants of the virus

Mutations in the novel coronavirus SARS-CoV2 are a major concern as they might lead to drug or vaccine resistance. Although most mutations may be detrimental to the virus's function, some will confer a significant advantage to the virus, helping it propagate and spread more rapidly. Many people infected with COVID-19, experience symptoms such as sneezing, which can lead to the virus spreading more widely.

In COVID-19, the SARS-CoV2 virus mutates about twice a month, almost half the common influenza virus's rate. Despite the reduced level of activity, tens of thousands of mutations continue to be documented, some of which lead to 'strains' or 'variants' that spread more aggressively in humans and result in hospitalization.

**Dr. Navaneeth Krishnamoorthy**, a molecular biochemist along with Dr. Khalid Fakhro, Chief Research Officer at Sidra Medicine, developed models of mutations in the main protein of the COVID-19 virus. The project is part of Sidra Medicine's Precision Medicine Program aimed at addressing questions related to what spots (sites) of the COVID-19 virus do not mutate and whether it is possible to map them and use them to guide vaccine design.

Dr. Navaneeth Krishnamoorthy, Molecular Biochemist at Sidra Medicine said: "It was fundamental that we addressed the question around mutation free spots, because genetic theory suggests that these spots are mostly conserved due to mutations that would be highly detrimental to the virus itself, i.e., they need to be conserved to ensure the virus can function properly."

Dr. Khalid Fakhro, Chief Research Officer at Sidra Medicine said: "New mutations are bad in two ways, firstly, the virus may become more harmful and more efficient at invading its host and spreading, like the prevalent variants in the UK and South Africa. Second, the virus may change the shape of the site recognized by antibodies, thereby enabling the virus to evade the human immune response and continue spreading even in vaccinated individuals."



Dr. Krishnamoorthy and Dr. Fakhro spent several months analyzing more than 19,000 mutations circulating worldwide in the SARS-CoV2 main viral protein (protease) to map the mutation 'coldspots'. They successfully identified a map of mutation 'coldspots' and concluded that these are ideal sites for targeting the coronavirus. Their findings were published by the international union of biochemistry and molecular biology (IUBMB), in their flagship journal, *Life*, giving it full attention on the April 2021 cover page.

Dr. Krishnamoorthy continued: "It is a futuristic approach because we found conserved regional patterns (among the coronavirus family) near the mutation-free spots that can be targeted effectively now and in the future when similar viruses emerge. The identification and short list of these coldspots offers a new perspective to target the SARS-CoV2 while avoiding mutation-based drug resistance. The study pinpointed optimum target sites and opened new avenues for the design of mutation-free antivirals. This important work is as timely as it is valuable, as major countries around the world are releasing and trialing vaccines and drugs or are in the process of approving anti-COVID-19 agents."

Dr. Fakhro concluded: "The fight against COVID-19 is constantly evolving. To resolve this global healthcare challenge in such critical times, we have to understand the viral defense mechanism of such mutations; our findings further this understanding and align with Sidra Medicine's ambition to deliver precision medicine."

## QF's Sidra Medicine research to help develop targeted therapies for childhood cancers

Precision medicine is an emerging healthcare treatment approach that offers personalized care, and is set to play a major role in the future of healthcare, particularly in complex diseases like cancer. The establishment of Sidra Medicine's pediatric cancer biorepository to develop personalized cancer therapies for pediatric patients is a big step towards this.

By tailoring treatment to fit each child, the focus will shift from treating a category that the child fits in, to treating the child's individual cancer in a very precise way. This approach will also ensure no child is being exposed to more chemotherapy or radiation therapy than is necessary thereby minimizing any side effects and toxicity.

**Dr. William Mifsud**, Attending Physician at Qatar Foundation's Sidra Medicine's Anatomic Pathology division, explains some studies have shown that particular chemotherapeutic agents can have an impact on cardiac function. "Through tailoring treatment, we want to strictly limit exposure of a child to such drugs to precisely the amount that is needed so we don't just treat the child but do so with minimal effect to their quality of life." In order to provide such tailored healthcare treatment, research begins at the biorepository.

"A biorepository is a place where biological samples are collected, processed and stored to support scientific research," said Dr. Wouter Hendrickx, Principal Investigator (PI) of the Pediatric Cancer Omics Lab at Sidra Medicine.

Every sample that goes into the biorepository will be an investment towards better pediatric cancer care. Analyzing diseased tumor tissue will allow researchers improved biological understanding of how pediatric cancers operate, including why some cancers respond or don't respond to a particular drug, or why some tumors grow much faster than others and why some recur.



**Dr. Hendrickx** explains that the idea to establish a national pediatric cancer biorepository was born three years ago when Sidra Medicine was recognized as the only center for pediatric Oncology in the country by the National Cancer Program in Qatar.

As pediatric cancer cases from all over the country were transferred to Sidra Medicine, the general consensus among the medical community was that this was a great opportunity to set up a number of research projects focused specifically on pediatric cancers, particularly with the aim of developing more personalized medicine for children with cancer in Qatar.

The lack of a local pediatric cancer biobank was one of the biggest hurdles to doing proper research on childhood cancers; it is noteworthy that Sidra Medicine has been able to change that with the establishment of the pediatric cancer biorepository.

The pediatric cancer biorepository is offering a chance at having an improved understanding of pediatric cancer specifically among Arab populations, and thereby bridging the gap between clinical research and effective cancer care.

## Qatar's Sidra Medicine: An oasis of science in the Middle East

More than 25 years ago, Her Highness Sheikha Moza bint Nasser had a vision: to transform Qatar from a mere oil- and natural gas-rich desert state into an oasis of knowledge. This vision was realized in 1995 through the establishment of the Qatar Foundation for Education, Science and Community Development. Fast forward to 2021, and Qatar's capital city Doha now boasts a vibrant intellectual community nurtured by the Qatar Foundation, replete with world-class universities, biomedical research centers, a women's and children's hospital, and more, all within walking distance.

"In terms of clinical expertise, we have a Who's Who across disciplines," explains Khalid Fakhro, Ph.D., Chief Research Officer at Sidra Medicine. "That creates a community, a knowledge economy, that builds sustainability for academic medicine in this region."

Sidra Medicine, whose campus is in the heart of Doha's Education City, is also a brainchild of that initial Qatar Foundation vision. Her Highness had always wanted a women's and children's hospital modeled after the leading centers around the world. Built with a vision emphasizing genomics and precision medicine in the treatment of pediatric cancers and rare diseases, it is now the country's largest tertiary care hospital for children and young people.

"We've come a long way in a short time, and for those interested in doing gene discovery and biomedical research in a Middle Eastern context, and investigating patients from this part of the world, Sidra Medicine is the place to be," says Fakhro.

### Three research pillars

Sidra Medicine was built from the ground up to integrate clinical, educational, and research aspects found at leading academic medical centers. Its research strategy stands on three pillars: patient biorepositories, advanced diagnostics, and personalized therapies.

When a pediatric cancer patient is referred to Sidra Medicine, their biopsy sample is exhaustively analyzed across different genomic and proteomic platforms, with the remainder safely stored for future examination. "Being basically the only center that treats pediatric cancer in the country means that you have perfect data. We have integrated genomic data,

anatomical data, and pathological data, all in an electronic format," explains Davide Bedognetti, M.D., Ph.D., director of the cancer research program.

For children with aggressive disease—whose tumors are relapsed or resistant to therapy—"we have an expedited clinical research pipeline in which we use whole-genome sequencing, whole-exome sequencing, RNA sequencing, and methylomics, to potentially discover targetable alterations," explains Bedognetti. "This information can be used to determine whether a patient might be eligible for a suitable drug that already exists."

### A case in point

"We don't just sequence the patient's genomes and identify mutations. We want to study the function of the genes so that we know what we can do for the patient," says Bernice Lo, Ph.D., a principal investigator studying immune dysregulation in children.

When two siblings with severe autoimmune disease were not being helped by traditional immunosuppressive therapies, their genomes were sequenced and found to have mutations in the LRBA gene. Lo's previous work had identified LRBA as a regulator of an important immune inhibitory molecule, CTL A4 (1), and abatacept—a CTL A4 mimic—was shown in the lab to compensate for the defective LRBA. Dr. Amel Hassan, Senior Attending Physician in Pediatric Allergy and Immunology, treated the patients with a personalized treatment regimen using abatacept, which resulted in significant improvements in their clinical condition, including reduced inflammation, fewer gut symptoms, and significant weight gain.

"By combining the clinical and molecular details, we can help clinicians decide on the proper treatment," Lo says. "The very tight integration between a clinic and research means that you can go from the important clinical questions to the bench, and then back to the bedside."

### Consanguinity

Sidra Medicine plays an important collaborative role in Qatar's national genome program, which is driven by another Qatar Foundation entity, the Qatar Genome Programme (QGP).



QGP provides sequencing for Qatari citizens and long-term residents (consented through partner biomedical institutes) to empower research studies into precision population health. The national reference genome lab and bioinformatics infrastructure supporting this ambitious vision are housed at Sidra Medicine.

Sidra Medicine's services are also on offer to other pediatric patients in the region. "When you look at the vastness of the greater Middle East and North Africa region, we have so much diversity here that has been underrepresented in global databases," Fakhro explains. Sidra Medicine is trying to remedy that by sequencing genomes, conducting and hosting clinical trials, and sharing data. "We are doing things in a localized context, but also serving our populations, our diaspora, and the rest of the world in the process."

There is also elevated consanguinity—shared genetic history—in the Arab population, which might favor the accumulation of homozygous recessive single-gene defects (two copies of the same unique recessive allele) that could lead to rare and often debilitating disorders. This creates opportunities for novel gene discovery, as consanguinity enables easier identification of these mutations. "In many cases, discovery of these genes sheds light on novel biological pathways that can be targeted by drugs or small molecules," says Fakhro.

### Cancer and the immune system

Cancer is the leading cause of childhood death by disease. Sidra Medicine is interested not only in how a tumor evolves, but also in how the immune system interacts with the tumor cells. "We know very little about the role of the immune system in controlling or promoting tumors in children," says Bedognetti. "Cancer immunotherapy has revolutionized the treatment of tumors in adults. But it's still in its infancy when it comes to pediatric tumors."

In research published in February 2021 in *Immunity*, Bedognetti and an international team, together with Elad Ziv, demonstrated that pre-existing anticancer immunity depends heavily on a patient's genetic background (2). "We analyzed the relationship between 11 million patient DNA germline variants from The Cancer Genome Atlas and the intensity of the immune response against the tumor," he relates. "About 25% of the immunologic parameters assessed, including those associated with responsiveness to immunotherapy, were influenced by the hosts' genetics. This understanding, and the identification of potential causal genes, may lead to more personalized immune therapies," says Bedognetti.

### Open for Business

Sidra Medicine is at an inflection point. Their Advanced Cell Therapy Core will soon boast a good manufacturing practice (GMP)-grade facility, enabling them to manufacture cell and gene therapies for their patients. It's also an important first step toward developing novel experimental drugs, biological products, and advanced therapies to foster new cures for diseases affecting the Qatari population, points out Fakhro. In addition, he says, "a company or institution can run its entire clinical trial here in a highly localized context."

Sidra Medicine welcomes collaboration, particularly with industry, and is seeking exceptional faculty and postdocs. As Fakhro puts it, "Here's an academic medical center sitting in the Middle East that has Middle Eastern phenotypes and genotypes. We have exceptional physicians, cutting-edge technologies, standard operating procedures, and resident experts across many multidisciplinary fields under the wider Qatar Foundation umbrella. There is always room for more innovation and advancement, especially in the growing field of precision medicine."

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## Italian Embassy Visits Sidra Medicine: Italian Research Day



On April 15 we celebrated Italian Research Day in collaboration with the Italian Embassy in Doha to promote excellence in scientific sectors. Innovation, competitiveness and research into technologies respondent to the needs of citizens. The campaign was to promote the growing number of collaborations and projects contributing to consolidate Qatar-Italy scientific relations. The Sidra Medicine Research team, which includes more than 35 different nationalities, is actively engaged in precision medicine, from improved diagnostics of rare genetic diseases to cancer research and new therapies, aiming to boost patients' immune responses through innovative approaches in personalised medicine.



## Carnegie Mellon University Qatar – Career Day



**Carnegie  
Mellon  
University  
Qatar**

Sidra Medicine participated in Carnegie Mellon University Qatar's (CMU-Q) Professional Services Career Day, providing its students with a unique opportunity to network and meet with leading professional services companies. The Career Development Office (CDO) is dedicated to connecting well-prepared, knowledgeable and enthusiastic students with potential employers. CDO is a bridge for students who are moving into the professional world, ensuring they have the workplace skills that complement their first-class Carnegie Mellon education. The Career Development Office opens the channels of communication so employers see the quality of CMU graduates, and students gain real-world experience.

## Precision Medicine and Functional Genomics Conference PMFG 2021



Sidra Medicine hosted the sixth edition of our flagship conference on precision medicine this year. It was hosted in Qatar, but was streamed worldwide with over 500 attendees from all over the world. The conference was led by a distinguished panel of Qatar-based subject-matter experts and institutional leaders. The conference concluded with the state of Precision Medicine in Qatar and the hope for the future. Opening remarks were given by Dr. Khalid Fakhro, Chief Research Officer; Dr. Ammira Akil, and Dr. Younes Mokrab, organising chairs.

The three-day conference dived into a deeper understanding of the themes of Genomic Medicine and Precision Care in Maternal and Child Health as well as Advances in Innovative Therapies provided by a host of eminent scientists and physicians, all leaders in their discipline.

Precision medicine is data and knowledge-driven, and a critical new tool in the fight against pediatric disorders. The Sidra Medicine Precision medicine program supports innovative pediatric genomic research that will yield lifesaving therapeutic options.

Medicine has been witnessing a drastic transformation to harness the huge amounts of data that is being obtained on patients and their genomes worldwide. Having in mind

the goal of helping catalyze such endeavor in the Arab and Middle Eastern region, in this year's PMFG21, some of the world's best experts and pioneers were gathered to talk about how data resources have been enabling genomic medicine, the impact of large sequencing consortia as well as the latest developments for genome and phenome actionability for healthcare.

A key message was that to improve outcomes of fertility, it is important to understand pre-implantation genetics, and that precision medicine starts as early as in-utero. Outstanding speakers shared their insights and examples of the translational and clinical application of precision medicine on multiple types of diseases.

This year PMFG2021 comprised an exciting program with the participation of outstanding speakers, both international and from the region. Sidra Medicine is a leading model for precision medicine in the region, and the scientific knowledge gained through the conference will help to converge the efforts into this goal.

The PMFG Chairs also thanked the conference partners—Hamad Bin Khalifa University, Hamad Medical Corporation, Qatar Biobank, Qatar Cancer Society, Qatar Genome, Qatar University, and Weill Cornell Medicine-Qatar.

## Sidra Medicine Research Day



The Research Day was held on Saturday, November 20th, from 8 a.m.–5 p.m., and attended by over 100 employees joining the meeting in the hospital building auditorium, overflow room, and via live stream. Dr. Khalid Fakhro, Chief Research Officer, started proceedings with a warm welcome speech, followed by interesting talks and lectures from our Research Department Clinical Directors, Clinical Researchers, Principal Investigators, and even Junior Trainees. International Keynote Speakers were also invited to the event and included Dr. Nancy Cox and Dr. Mustafa Khokha from the United States. Dr. Nancy Cox is the Director of Vanderbilt Genetics Institute and Division of Genetic Medicine and a Mary Phillips Edmonds Gray Professor of Genetics. Dr. Cox is a quantitative human geneticist with a long-standing research program in identifying and characterizing the genetic component to common human diseases. Her current research focuses on large-scale genomic integration with other “-omics” data and biobank electronic medical records data. The other keynote speaker, Dr. Mustafa Khokha, is a Professor of Pediatrics and Genetics at Yale University School of Medicine. He completed his pediatrics and pediatric critical care training at Washington University and UCSF.

His research interest includes the discovery of candidate genes from patients with birth defects and other critical illnesses. There was also a poster contest during the

event, which saw submissions from Clinical Researchers in different departments, and Senior and Junior Scientists. A total of 80 posters competed, and the winners were chosen in various categories. The judging panel consisted of the International Speakers and Clinical Directors. Clinical Researcher winners included Tariq Wani, Sanoj K.M.Ali, Samir Gupta, Sanaa Sharari, Zenab Siddig, and Shabir Moosa. Saroja Kotegar Bakayya, Rozaimi M. Razali, Jessica Roelands, and Selma Maacha won in the Senior Scientists category. The Junior Scientist winners were Ahmad A. Al Shaibi, Shima Sherif, Wehedy Eman, Mohammed Toufiq, Mona Abdi, and Mohamed Alsabbagh. All posters displayed at the event spoke volumes to the competence and creativity of Sidra Medicine Researchers; they do think outside the box. To wrap up the program, Dr. Fakhro, Dr. Cox, and Dr. Khokha handed out the awards for the best poster plus the awards for honorable and dedicated employees of Sidra Medicine Research Department nominated by their line managers and their colleagues.

The award categories were Core Exceptional Service, Core Innovation, Core Excellence, Research Coordinators Excellence, Admin-Extreme Dedication, Admin-“Protector of the Realm,” Admin-“Mountain Mover,” Outstanding RS, Research Specialist Extreme Dedication, Outstanding Senior Scientist, and Senior Scientist Extreme Dedication.

## BREAKOUT STORY

# Monitoring Evolution of Inflammatory Bowel Disease to Predict Cancer Risk



More than 6.8 million people worldwide are estimated to be living with inflammatory bowel disease (IBD), an inflammation or swelling in the gastrointestinal tract caused by genetic and environmental factors. IBD is becoming more prevalent in the pediatric population in Qatar as the global trend shows. Between 1990 and 2017, there was an increase of 85.1% of IBD patients worldwide.

**Dr. Mamoun Elawad**, Chief of Pediatric Gastroenterology and Director of inflammatory bowel disorder, has been receiving more pediatric patients with symptoms of IBD each year. According to him, there are about 172 pediatric patients and 2400 adult patients with IBD in Qatar. IBD is becoming more common among Qatari population due to migration, changes in diet and environment. In 2020, there were about 36 new pediatric patients with IBD and about 48 adult patients diagnosed with IBD at Sidra Medicine.

IBD cases are increasing annually, and parents of pediatric patients visit Sidra Medicine for help. Therefore, as a leading academic medical centers in Qatar, Sidra Medicine is investing in research projects that require tight collaboration between researchers and clinicians to provide personalized medicine to each patient.

“Personalized treatment for inflammatory bowel disease is crucial because clinicians have to look for clues as to which type of treatment that best works for each particular patient with particular disease. The different methods of Precision Medicine will allow clinicians to prescribe the right treatment for the right person from the beginning rather than trying different medicine before getting the right medicine that suits a particular patient,” said Dr. Elawad.

Dr. Elawad reached out to Dr. Cristina Maccalli, Principal Investigator at the Laboratory of Immune and Biological Therapy, to collaborate on a study of evolution of IBD to track its progression towards malignancy and risk to develop colorectal cancer. Clinical and research team from Sidra Medicine along with collaborators from one of the leading national medical centers are developing a study aimed at obtaining a deep, comprehensive genomic, molecular, and immunological characterization of patients to identify the markers that can predict evolution of a disease, capture patients’ pathophysiological heterogeneity and classify disease subsets.

“The ultimate goal is to develop a Precision Medicine program for the monitoring patients to predict the evolution of the disease towards severe grades and to predict the risk of developing colorectal cancer,” said Dr. Maccalli.

Further investigation on evolution of IBD to predict cancer risk is ongoing. Integration of data from multi omics platforms that are currently under investigation in Dr. Maccalli’s team, discovering general molecular insights in IBD patients and the evolution of disease. With current findings, present and future IBD patients at Sidra Medicine will be able to access personalized treatment based on their conditions promptly with no delays in identifying markers that predict the evolution of the disease.

## BREAKOUT STORY

# Sidra Medicine Using Art Therapy and Museums to Improve Adolescent Mental Health



Medicines, body scans and intensive procedures are common and necessary treatments in a hospital environment. However, Sidra Medicine takes a holistic approach for patient care—this time through art therapy and museums.

In collaboration with Weill Cornell Medicine in Qatar and the National Museum of Qatar, Sidra Medicine is part of a study awarded by Qatar National Research Fund to build on existing art therapy practices. Michelle Dixon, Art Therapist at Sidra Medicine, is serving as co-principal investigator with **Dr. Finza Latif**, Program Director for Sidra Medicine's ACGME-I accredited Child and Adolescent Psychiatry Fellowship to facilitate the online art therapy sessions of the project. These telehealth sessions will involve Sidra Medicine patients between the ages of 14–17 diagnosed with anxiety and depression.

For the past four years, Ms. Dixon has been holding bedside and waiting room art therapy sessions with the child and adolescent patients at Sidra Medicine. She uses her counselling background to support children in using art materials with a therapeutic perspective. Not only does this foster their creativity, but it also encourages them to talk about their feelings. “The hospital can be a traumatic event for a child and art therapy normalizes it into a nonthreatening space,” said Ms. Dixon. The impact of art therapy also stems from the sense of control young patients can reclaim through it. Ms. Dixon presents them with various art materials to choose from, all of which can serve a different purpose for the patient.

Sculpting with clay and slime, for example, can be good for connecting with their body, experiencing emotional regulation, and practicing breathing activities. Collage

and drawing allow more control and making symbols in artwork can enable people to think about their life from a different perspective. Moreover, if a child is quiet and withdrawn, they can listen to music and be given more space while working on their art. Just like treatments can be personalized through Precision Medicine, art therapy can also be curated to the needs and personality of the patient through informal assessments.

The art therapy research project will enable Sidra Medicine to build on its existing art therapy practices. The group setting of the telehealth sessions will be particularly helpful in the context of COVID-19 as adolescents have not had the same lifestyle due to social-distancing restrictions and school closures. The hospital has already seen the benefits of art therapy, within a telehealth context, in encouraging connectivity with others.

With NMoQ providing access to its archives and artefacts, participants will also be learning about Qatar's local culture, history and stories. This will provide Sidra Medicine with a more extensive and unique evidence base of what resources work well in art therapy, continued Ms. Dixon.

“There are currently no robust empirical studies of Art Therapy in the Gulf region and only a small number of countries in the world have produced research on telehealth in art therapy and the role of art museums in improving wellness factors,” said Dr. Alan Weber, Professor at Weill Cornell Medicine-Qatar and lead Principal Investigator of the study. “Thus, Qatar and Sidra Medicine will become recognized international leaders in this field.”

**Written by Areesha Lodhi**





PUBLICATIONS  
REGISTRY

# PUBLICATIONS REGISTRY

## CANCER

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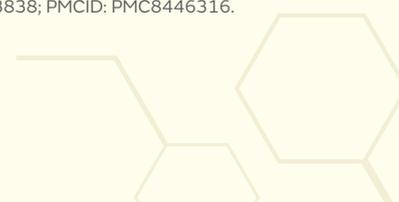
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...2021 ultimately became one of the most productive and innovative years at the Research Branch. We owe a most sincere gratitude to our hard-working research staff... to our diverse clinical collaborators... and to all the families and patients who entrust Sidra Medicine to deliver the highest-quality, research-driven care.

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