

2024 ANNUAL REPORT

SIDRA MEDICINE RESEARCH



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TABLE OF CONTENTS







03

WELCOME NOTE BY CRO



Prof. Khalid A. Fakhro Chief Research Officer

As we reflect on the past year, we take immense pride in the transformative strides made in research and innovation at Sidra Medicine. Our commitment to advancing translational research promises to position Qatar as a global leader in research-guided care for women and children. In fact, precision medicine can only reach its true potential when patients become the center of everything we do — whether it's using WGS to end a diagnostic odyssey, or a novel gene therapy to cure someone — and every patient's journey is enriched through research.

Amongst all achievements you will find in the 2024 Annual Report, several stand out. First, the Research Branch underwent a thorough strategy refresh, paving the way for a new Research Strategic Plan for 2025-30. This process not only helped us map current areas of strength, but also identify key priorities for the future. These include doubling down on our investment in genomics as a key diagnostic and preventative screening technology, commiting to building clinical research and trials capacity for rare and orphan diseases, and expanding our scope to embark on implementation and health services research – the latter promising to transform Sidra Medicine into the region's leading academic learning health system.

An important aspect to executing on the new strategy is developing an ecosystem that supports innovation and encourages capacity building. In terms of scientific productivity, our commitment to rigorous, high-qualithy discovery remains as high as ever. In 2024, our scientists published 97 papers (mean IF - 8.7), were awarded 6.7 M QAR of grants, mentored 69 Masters and PhD students, and hosted 12 visiting scientists and externs across their labs. In terms of support for clinical activities, our core facilities have been intimately involved in launching clinical grade sequencing, long-read sequencing, cord blood banking and, most recently, supporting a pioneering BMT program at Sidra. These efforts not only demonstrate our world-class capabilities, but for the first time provide local options for services which used to require families travel thousands of miles for treatment.

Finally, we are very proud of our efforts in thought leadership, as our faculty are internationally recognized in their fields, and our exceptional conferences and workshops, and training programs are very well-attended. In 2024, we successfully launched Sidra's Science Café, which promises to improve public engagement and undersanding of science, and has the power to inspire the next generation of scientists.

In conclusion, as we embark on the next phase of our journey, we remain committed to excellence, collaboration, and impact-driven science. We extend our heartfelt gratitude to our trainees, researchers, clinicians, and partners who have played a vital role in driving our vision forward, and to hospital leadership for their exceptional support of Sidra's academic mission to improve patient lives. Together, we will continue to push the boundaries of science and medicine, and bring the cutting-edge of research-driven healthcare to our patients and their families.

LEADERSHIP STRUCTURE

OUR MISSION AND STRATEGY

Operating Model

To achieve its potential, Research at Sidra Medicine encompasses four complementary streams - diagnostic, therapeutic, and implementation - which together combine to achieve an operating model in line with leading ALHS worldwide, as follows: Precision Diagnostics (Dx) – Discovering why patients get sick through advanced diagnostics & screening.

Personalized Therapies (Tx) – Improving patient outcomes through bedside research and clinicial trials.

Implementation Research (Ix) - Measuring impact on health services and quality improvement of Dx and Tx above. World-class Ecosystem (Ex) - Positioning SM as regional leader for alliances/partnerships and biomedical talent.

Virtuous Cycle of Precision Medicine



These priorities and implementation are underpinned by a strong culture of innovation, and recognition of our intellectual property and technical know - how.

Our Strategic Priorities for 2024-2029

- Understand the mechanistic basis of rare and complex diseases and develop preventive screening.
- · Improve patient outcomes through clinical research and advanced therapies.
- Enhance the effectiveness and experience of patient care.
- · Create a world-class translational research environment attracting global talent and industry and academic partnerships.





Our Mission

We deliver leading clinical and translational research of value to our patients, the State of Qatar and the world.



Our Goal

Women and children living with rare and complex diseases in Qatar and travelling to Qatar achieve improved health outcomes aided by cuttingedge translational research and genome-informed patient care.



Our Values Trust, Transparency, Teamwork, Care, Innovation and Efficiency.

INTERNAL RESEARCH COUNCIL

The Internal Research Council (IRC) at Sidra Medicine serves as the key governance body responsible for overseeing the institution's research roadmap. Its primary objective is to ensure that our research remains internationally competitive while adhering to necessary governance and assurance frameworks that support effective functioning and delivery of the roadmap. Through strategic planning, rigorous oversight, and fostering collaborative efforts, the IRC has worked to align research initiatives with organizational priorities.

Achievements and Activities Updated 5-Year Research Strategy Activation of Translational **Research Programs Oversight of all Research** Studies & Clinical Trails Awarding of Competitive Internal Research Funding (IRF 2024)

Impact and Contributions The IRC's efforts have laid a robust foundation for advancing clinical research at Sidra Medicine. By establishing clear guidelines, activating key programs, and fostering collaboration across disciplines, the Council has enhanced the alignment of research initiatives with our strategic goals, improved the clarity and focus of clinical research programs through the adoption of SMART Goals, and ensured that research efforts remain competitive on an international scale

Internal Research Council Membership List





Dr. Ibrahim Janahi Division Chief, Pulmonology Senior Clinician Scientist Vice Chairperson



Dr. Sonia Davila Executive Director, Translational M Senior Research Scientist Member



Dr. Chiara Cugno Director, Advance Cell Therapy Co Senior Research Scientist Member



Dr. Johnny Awwad Division Chief, Reproductive Medicin Senior Clinician Scientist Member while adhering to governance frameworks. The IRC has made substantial strides in advancing Sidra Medicine's research mission through meticulous planning, collaboration, and oversight. The initiatives undertaken this year position Sidra Medicine to achieve transformative breakthroughs in healthcare and reinforce its reputation as a leader in clinical research. The Council remains dedicated to fostering innovation and excellence in research in the coming years.

Prof. Khalid Fakhro Chief Research Officer Chairperson



Muhammad Ali Hameed Executive Director, Research Core Facilities Senior Research Scientist Member



Dr. Mamoun Elawad Division Chief, Gastroenterology Senior Clinician Scientist Member



Dr. Sandra Holmes Director, Nursing Education Prof Practice & Standards, Nursing Member



Dr. Tariq Abbas Senior Attending Physician, Urology Senior Clinician Scientist Member

SIDRA MEDICINE: OUR RESEARCH AT A GLANCE

37 Nationalities at Sidra Medicine Research



206 Total Staff 143 Female 18 6 47 106 . Research Operations Team . Office Management Team . Researchers in Core Facilities . Researchers in Translational Medicine . Grant Funded







Texas A&M University - Qatar Qatar Research Development and Innovation Council (QRDI) Qatar University (QU) Hamad Bin Khalifa University (HBKU) Qatar Museums (IN-Q) Weill Cornell Medicine -Qatar (WCM-Q) Qatar Bio Bank (QBB) The Cultural Village Foundation -Katara University of Doha and Science and Technology (UDST) Qatar Cancer Society (QCS)



PUBLICATIONS







SCIENTIFIC DIVISIONS

Dr. Sonia Davila joined Sidra Medicine in December 2023 as Executive Director - Translational Medicine. She completed her undergraduate and graduate studies in Spain. Upon graduation she moved to Yale University and in 2004 Sonia joined the Genome Institute of Singapore as a postdoctoral fellow on nephrogenomics. She was rapidly promoted to Research Scientist and to Principal Investigator. Her work focused on human genetic susceptibility to infectious diseases. In 2016 she started working at SingHealth-Duke NUS AMC and was Deputy Director of Precision Medicine (PRISM) and Director of Research at their Genomic Medicine Centre.

TRANSLATIONAL MEDICINE

The Translational Medicine department plays a vital role in advancing Sidra Medicine Research's vision, paving the way for Sidra Medicine to become a premier academic medical center and a leading destination for patients seeking cutting-edge medical care in the region. The translational medicine department is dedicated to the development and implementation of precision medicine approaches aiming to deliver accurate molecular diagnosis that guide effective therapeutic decisions for our patients.





Sidra Medicine's commitment to innovation in precision medicine is reflected in the work of its research groups, which leverage high-throughput profiling technologies. The Translational Medicine department at Sidra Medicine is a cornerstone of the institution's research strategy. Through its focus on precision medicine, innovative research, and global collaboration, it is poised to make significant contributions to the field of medicine and improve patient outcomes.



Ammira Al-Shabeeb Akil, PhD

Laboratory of Precision OMICs Research and Translational Medicine (PORT Lab)

Bio:

Dr. Akil holds a leadership role at Sidra Medicine's Metabolic and Mendelian Clinical Genomic Research Program and serves as Lead Principal Investigator for the Precision OMICs Research & Translational Science initiative in Doha, Qatar. She partakes in extensive contributions to clinical genomic research, particularly in rare and complex metabolic diseases. Dr. Akil secured over \$7M in research funding from prestigious national and international organizations, strengthened Sidra Medicine's partnerships with the biotechnology and pharmaceutical industries, and made significant contributions to capacity building by training and mentoring hundreds of externs, equipping the next generation of researchers and clinicians with cutting-edge expertise in genomic medicine. She is an active member of several prestigious organizations, including the American Diabetes Association, ISPAD, IDS, EASD, and INNODIA.

Lab Interest:

Dr. Akil's research focuses on advancing the integration of genomic technologies into precision medicine, particularly in newborn screening, metabolic, and Mendelian disorders. A key element of her work involves discovering innovative research ideas and transforming them into successful funding applications and translational research. Dr. Akil's lab collaborates actively with leading scientists and global partners to explore novel genomic and multiomics solutions to ensure that our research remains at the forefront of scientific discovery while also addressing critical health challenges. The Akil lab interests extend to populationbased multi-omics research, integrating genomic, transcriptomic, proteomic, and metabolomic data to unravel complex biological mechanisms underlying human diseases. Additionally, they focus on leveraging genetic and polygenic risk scores for predictive diagnostics, particularly in type 1 and type 2 diabetes.



Ajaz Ahmad Bhat, Reem Al-Karbi, Ammira Akil, Sura Ahmed Hussein, Ikhlak Ahmed.



Dr. Terranegra is the Principal Investigator of the Laboratory of Precision Nutrition. She 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. Annalisa achieved strong experience in molecular biology and genetics during her career at the University of Milan, Italy. Before joining Sidra Medicine in 2014, Dr. Terranegra covered a consultant position in genetics at San Raffaele Hospital, Milan, Italy and a research fellow position in nutrition at San Paolo Hospital. Dr. Terranegra also has extensive teaching experience as an Assistant Professor and Lecturer at University of Milan, Italy.



Salma Hayder, Shalima Lathief, Arun Lakshmanan, Annalisa Terranegra, Ayaaz Ahmed, Fatima Mohamad, Dena Al Dasoogi, Surya Devarajan.

Annalisa Terranegra, PhD Laboratory of Precision Nutrition

Lab Interest:

The Laboratory of Precision Nutrition (PreNutri Lab) at Sidra Medicine focuses on characterizing the mechanisms underlying the role of food and nutrients in the development and the management of pregnancy complications, infertility, and pediatric non-communicable diseases. The PreNutri lab research projects focus on understanding the effect of diet on gut microbiota and epigenetic mechanisms, as well as diet interaction with the genomic background, using the most advanced technologies, such as bacterial 16S rRNA-sequencing, DNAmethylation arrays, DNA genotyping and nutritional assessment. Thanks to local and international collaborations, the PreNutri lab is currently running many studies on female reproductive health, perturbation of the motherbaby axis during the pregnancy and early baby life, pediatric type 1 diabetes, adult and pediatric obesity, cardiovascular diseases, inflammatory bowel diseases, and nephrotic syndrome.



Bernice Lo, PhD

Laboratory of Immunoregulation

Bio:

Dr. Bernice Lo is the Principal Investigator of the Laboratory of Immunoregulation. She is also a Joint Assistant Professor at HBKU. Bernice 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.

Lab Interest:

The research focus of Dr. Lo's laboratory is on understanding the molecular mechanisms of immune regulation and tolerance. Her approach includes identifying the genetic basis of disease in patients with immune dysregulation and autoimmunity with the ultimate goal of elucidating the molecular pathways involved and, therefore, revealing new therapeutic targets for disease treatment or novel drugs for immunomodulation. Dr. Lo's lab specializes in using cellular and biochemical techniques to uncover the role of newly discovered gene mutations in causing disease and in understanding the function of these genes in immune regulation. Her research interests also include identifying modifier genes and elucidating their functional role in immunologic disorders with incomplete penetrance. She is also interested in developing methods for disease diagnosis or evaluating treatment efficacy by assessing immune biomarkers.



Bio:

Dr. Abdelalim is a Principal Investigator at Sidra Medicine, leading the Laboratory of Pluripotent Stem Cell Disease Modeling. He earned his PhD in Medical Science from Shiga University of Medical Science in Japan then became Assistant Professor there. Before joining Sidra Medicine, he held the position of Senior Scientist at QBRI until February 2024. Notably, he established Qatar's first pluripotent stem cell program focused to diabetes research. Dr. Abdelalim also serves on the editorial boards and has authored over 75 peerreviewed articles, received numerous research grants and honors for his contributions to the field. Dr. Abdelalim also holds a joint appointment as Associate Professor at HBKU, where he teaches Stem Cell Biology and mentors graduate students.



Katherine Whitmore, Yasmin El Bsat, Bernice Lo, Jonamae Dioso, Rafah Mackeh, Ameera Sadoun, Abdulrahman Ahmed Al Subaiey, Asha Elmi, Satanay Zuhair, Maryiam Osman.



AlDana Alnesf, Essam Mohamed, Yusra Manzoor, Noura Aldous, Ahmed Kamel, Karim Esmat, Idil Ibrahim, Nafisa Fadulelmula, Simona Semaan, Tara Fathi, Hafsa Madni.

Essam M. Abdelalim, PhD Laboratory of Pluripotent Stem

Lab Interest:

Our laboratory is dedicated to unraveling the intricate disease mechanisms underlying diabetes and rare genetic disorders using human pluripotent stem cells (hPSCs). Leveraging iPSC technology alongside state-of-the-art gene editing and omics approaches, we strive to dissect the molecular intricacies of these diseases. By collaborating closely with clinicians at Sidra Medicine, we have unique access to patients presenting specific mutations or variants implicated in these conditions. Our multidisciplinary approach enables a deeper understanding of the molecular drivers behind these conditions. Ultimately, our research endeavors focus on identifying novel therapeutic targets, facilitating the development of precision treatments tailored to meet individual patient needs and making significant contributions to the field of personalized healthcare and regenerative medicine.



Khalid A. Fakhro PhD

Laboratory of Human Genetics and Genomics

Bio:

Prof. Khalid Fakhro is the Chief of Research and Chair of the Precision Medicine Program at Sidra Medicine, the largest tertiary care women and children hospital in Qatar. In addition to research and hospital duties, Prof. Fakhro serves multiple leadership roles in Qatar's growing biomedical ecosystem, including as a Board Member of the Qatar Precision Medicine Institute and Adjunct Professor at both Weill-Cornell Medical College and Hamad Bin Khalifa University, where he teaches and supervises Master's and Ph.D. students in genomics and precision medicine.

Lab Interest:

Prof. Fakhro leads the Laboratory of Human Genetics and Genomics, 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 publishing landmark studies on population structure, genome structural variation, and the role of Islamic ethics in genome research. To date, his lab has been awarded competitive grants exceeding \$8m to study genome structure and the genetic etiology of rare diseases and Autism, and their discoveries have featured in high-impact journals worldwide. Our laboratory is interested in interpreting the human genome (the instruction book of life).



Luis R. Saraiva, PhD Laboratory of Disease Modeling and Therapeutics

Bio:

Dr. Luis join Sidra Medicine in 2015 and currently is the Principal Investigator leading the Human Disease Modeling and Therapeutics Laboratory and the Congenital Malformations Clinical Research Program. Luis' academic journey took him to Harvard Medical School as a visiting scientist, funded by the Boehringer Ingelheim Fonds, followed by his first postdoctoral appointment at the Fred Hutchinson Cancer Research Center (USA). He served as Adjunct Faculty at the Monell Chemical Senses Center (USA) from 2016-2023, has been a Joint Professor at Hamad bin Khalifa University (Qatar) since 2018, and an Associate Professor Adjunct at Yale University (USA) since 2024.



Shima Salim, Amal Ibrahim, Khalid Fakhro, Rulan Shaath, Alya Al Kurbi, Geethanjali, Najeeb Syed, Waleed Aamer, Aljazi Al-Maraghi, Elbay Aliyev.



Reem Hasnah, Kholoud Al Shafai, Luis Saraiva, Melanie Makhlouf, Diogo Manoel, Asma Al Naama.

22 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

Lab Interest:

Our laboratory develops and deploys biological assays to understand how specific genetic variants can cause the dysregulation of specific molecular mechanisms leading to disease. We collaborate with clinicians and leverage cell and animal models to apply an integrative strategy combining molecular biology, genetics, behavior, and omics techniques. Ultimately, we aim to advance fundamental science and translate our findings into new diagnostic and treatment options for patients suffering from rare genetic diseases. We also play a pivotal role in the implementation and advancement of the Congenital Malformations in Clinical Research Program at Sidra Medicine. Our efforts support clinical colleagues in gathering and analyzing patient cohorts with a diverse range of congenital malformations, with a particular focus on hypospadias, spina bifida, orofacial clefts, and congenital heart disease.

Matteo A Avella, Ph.D

Laboratory of Reproductive Biology

Bio:

Dr. Matteo Avella is a Principal Investigator in the Division of Maternal and Child Health at Sidra Medicine and is head of the Lab of Reproductive Biology. Prior to joining Sidra Medicine, Dr. Avella was an Assistant Professor in the Department of Biological Science at the University of Tulsa, USA and the School of Health Professions at Eastern Virginia Medical School (Norfolk, VA, USA). Dr. Avella completed his Ph.D. at the Polytechnic University of Marche (Ancona, Italy), studying the intestinal microbiota's effects on fish's early development and reproduction. Before his first academic appointment at the University of Tulsa, Dr. Avella received training as a human embryologist at Shady Grove Fertility (Chesterbrook, PA, USA).

Lab Interest:

Dr. Avella's lab investigates the mechanisms regulating gamete interactions, fertilization, and early embryonic development. A series of carefully orchestrated molecular events ensures monospermic fertilization, essential for successful embryonic development and a healthy pregnancy outcome. In collaboration with the Division of Genetics and the IVF program, we use next-generation sequencing to identify novel genes or variants in infertile patients from the Qatari population. In our lab, we use different model systems (e.g., zebrafish, mouse, and cell lines) to functionally characterize such variants or genes. In addition, we translate our discoveries to establish novel assays for assisted reproductive technologies. A precise characterization of the mechanisms regulating reproduction is instrumental in providing targeted fertility treatments to people requiring assisted reproductive technologies in Qatar.



Suma Garibova, Manar Mahmoud, Maha Abdulla, Matteo Alessandro Avella, Shoaib Nawaz.



Souhaila Al Khodor, PhD Laboratory of Microbiome and Biomarkers Discovery

Bio:

Dr. Al Khodor is Principal Investigator of the Laboratory of Microbiome and Biomarkers Discovery. She is an adjunct Faculty at the College of Health & Life Sciences in Hamad Bin Khalifa University. Souhaila has over 77 peerreviewed publications. She currently serves as the Section Editor for Metagenomics in the Journal of Translational Medicine and a speciality chief editor at Frontiers journal. Souhaila received her second master's degree and PhD in Microbiology and Immunology from the University of Louisville, Louisville, KY, USA. Before joining Sidra Medicine, Dr. Al Khodor worked as a postdoctoral fellow in the Signaling systems Unit, laboratory of Systems Biology, at the National Institute of Allergy and infectious Diseases (NIAID), National Institutes of Health (NIH) in Maryland, USA.



Mohamed Nadhir, Manoj Kumar, Selvasankar Murugesan.

Lab Interest:

Dr. Al Khodor's laboratory at Sidra Medicine focuses on using multi-omics (metagenomics, transcriptomics, proteomics) and computational biology aiming to understand the molecular mechanisms underlying various diseases. Her primary goal is to identify early biomarkers for disease prediction. With a special focus on pregnancy complications (mainly preterm birth and Gestational Diabetes Mellitus) and complex pediatric disorders such as Inflammatory Bowel Disease, her research at Sidra Medicine has attracted around 17 million QAR in external funds. Al Khodor's lab uses omics and computational biology aiming to understand the role of the microbiome and the complex microbial-immune cross-talks in pregnancy and chronic diseases.

Parul Singh, Samar Yahiya, Souhaila Al Khodor, Daliya Abubakar, Marwa Saadaoui, Duaa Ahmed, Ghada Yousif,



Wouter R.L. Hendrickx, PhD

Laboratory of Tumor Biology and Immunology

Bio:

Dr. Wouter Hendrickx is an investigator in Population Genomic Medicine Division of the Research Branch and member of the Cancer Program at Sidra Medicine. He Leads the Tumor Biology and Immunology Laboratory and has experience in stem cell and cancer research at the universities of Brussels (VUB), Leuven (KUL) and Norwich (UEA) where he gained a PhD in Medical Science. He has experience with classic molecular biology techniques as well as advanced 3D cell culture and proteomics technology. At Sidra Medicine he has focused since 2014 on the tumor immune micro environment deploying bioinformatic tools to analyze tumors for immune related signatures and other determinants of the immune phenotype and translating the findings to the wet lab environment.

Lab Interest:

We investigate the immunogenetic profile of colon cancer patients as Part of a JSREP funded grant. This is the first study of its kind in Qatar that enabled us to establish a Next Generation Sequencing (NGS) cancer cohort locally. In addition, we are involved in several international collaborations focusing on the therapeutic manipulation of the tumor immune microenvironment and the genomic determinations of these interventions. Following up on the genetic profiling of cancer samples, our wet lab research extends to functional omics to unravel the mechanistic basis of the NGS findings. For this purpose, we employ various methodologies, including 3D in vitro modeling and single cell analyses.



Rania Alanany, Shimaa Mohammed, Ayesha Jabeen, Wouter Hendrickx, Apryl Sanchez, Eiman Ahmed, Yara Elzeini, Christophe Raynaud.



Younes Mokrab, PhD

Laboratory of Medical and Population Genomics

Bio:

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. Dr Mokrab obtained a PhD in Bioinformatics from Prof. Tom Blundell lab, University of Cambridge, UK (2007). Upon joining Sidra Medicine, 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 conferences including Sidra Medicine Functional Genomics and CUDOS and holds academic appointments at WCM-Q and HBKU. Dr Mokrab is a co-author of > 45 publications in top tier including Nature and Cell journals and is the recipient of multiple awards and external grants (> 9.1 m QAR).



Abouelhassan, Ilhame Diboun, Haroon Naeem, Sondoss Hassan.

Lab Interest:

Dr. Mokrab's lab uses advanced wet and dry lab methods to study large-scale genomic and health records including EHR and Imaging to explore finescale population structure, its impact on disease risk, and the genetic architecture of both rare and complex diseases. His lab has a particular interest in Middle Eastern populations where high level of parental relatedness (consanguinity) is common, notably from Qatar and Arabia. In this regard, he has been working closely with the Qatar Genome Program, Genomics England, TCGA and other international consortia. Furthermore, he is leading a national effort to unravel the genetic architecture of rare neurodevelopmental disorders including epilepsy and the establishment of a national registry and disease cohort.

Radi Farhad, Melissa Tauro, Shabir Moosa, Zenab Siddig, Mohammadmersad, Younes Mokrab, Rawan

CORE FACILITIES

Muhammad Hameed is the Executive Director of Sidra Medicine's Research Core Facilities. He has over 15 years of experience in finance and business management across healthcare, energy, manufacturing, and education. His leadership experience has encompassed largescale operations in clinical research as well as core lab and biobank management, while successfully commercializing clinical services and spearheading initiatives to advance research and biotech innovation. He holds a Master of Business Administration from York University and a Bachelor of Engineering from the University of Windsor and is a Chartered Professional Accountant, Professional Engineer, Project Management Professional, and Certified Lean Six Sigma Black Belt.

The Advanced Research Core Facilities at Sidra Medicine's Research Department aim to promote a synergy between research and clinical care. Each of our core facilities houses cutting-edge equipment and technology and is staffed by top researchers. The multidisciplinary core facilities at Sidra Medicine promote collaboration between researchers and clinicians, fostering a holistic approach to patient care. Research Investigators and healthcare providers



can work closely with experts at the core facilities to deliver the best patient care possible and lead to a better understanding of diseases affecting Qatar's women and children. This synergy leads to advanced diagnostics, improved treatments, access to experimental therapies, and a culture of continuous improvement in medical practice. These benefits result in better patient outcomes and higher healthcare quality.



Omics Core Facility

The Omics Core Facility at Sidra Medicine serves as a hub to support research projects as well as international collaborations for translational and precision medicine. The Omics Core promotes world-class biomedical research by ensuring availability of the highest quality biospecimens and by offering a variety of services that complement the off¬ering of CGL Core, playing a critical role in accelerating discoveries in medicine.



Alia Al Massih, Rebecca Mathew, Sara Tomei, Iman Al Azwani, Muna Al-Hashmi, Santhi Raveendran.

Services include and are not limited to sample storage and tracking, DNA and RNA isolation from different sample types, longread sequencing (PacBio and ONT), optical genome mapping (Bionano Genomics), Illumina arrays (genotyping and methylation), Sanger sequencing, and High-throughput qPCR-based gene expression and genotyping profiling.

Clinical Genomics Laboratory

Sidra Medicine Clinical Genomics Laboratory (CGL) onffers access to a medium-to high throughput library preparation and sequencing methods. CGL employs the widely used and industry-leading Illumina Novaseq X Plus, Novaseq 6000, Nextseq sequencers complemented with Hamilton Robotics automation platforms and cutting-edge analytical instruments for sample quality control. CGL employs the widely used and industry-leading Illumina Novaseq X Plus, CAP accredited clinical grade whole-genome sequencing with 50-fold coverage of the human genome on Illumina Novaseq 6000, Nextseq sequencers, complemented by Hamilton Robotics automation platforms and innovative analytical instruments for sample quality control. We also offer cost-efficient 3' differential gene expression pro-filing and small RNA sequencing using Nextseq 500, and ultra-low input RNAseq for enriched cells and rare samples.

Zebrafish Functional Genomics Core

The facility employs the zebrafish as a valuable experimental model, offering a path toward advancing precision medicine. Our platform has achieved remarkable success in generating patient-specifinc genotype models. Our platform provides access to state-of-the-art technologies and equipment to study the genetic variation impact on cell, tissue, organ, and whole organism levels. Working with researchers and clinicians, the functional validation in the



Guishuang Wang, Kun Wang, Oleksandr Soloviov, Li Liu, Lisa Sara Mathew, Jodelyn Asinas, Li Wang.

Waseem Hasan, Sahar Da'as, Doua Abdelrahman, Tala Abu Arja.

zebrafish model sheds light on gene discoveries, clinical presentations, disease prognosis, and treatment strategies. We contribute to interpreting the Qatar Genome Project's unique findings and offer pediatric disease models for neurological disorders, cardiovascular diseases, congenital anomalies, and other diseases. Our support services encompass animal husbandry, training programs, and consultation for tailored research needs.

Advanced Cell Therapy Core

The Advanced Cell Therapy Core (ACTC) plays an important bridging function between the Research and Clinical Departments allowing the delivery of personalized, advanced and/or experimental therapies, including cell and gene therapy, to patient cohorts. Sidra Medicine has the first Good Manufacturing Practice Facility for cell and gene therapy in Qatar and is set up to provide advanced and experimental treatments. R&D activities go hand in hand with commercial manufacturing, enabling revenue generation in a virtuous circle. All ACTC personnel providing clinical services are QCHP licensed. Key activities are represented by private stem cell banking for innovative use of cord blood stem cells, manufacturing of experimental (i.e., gene-edited hypo-immune pancreatic islets for type 1 diabetes, CAR-T cells, etc.), and possibly marketed gene therapy products, manufacturing and biobanking of mesenchymal stromal cells for multiple clinical applications, and regenerative medicine.

Advanced and Novel Therapies and Treatments

Advanced Cell Therapy Core offers translational research from "bench to bed" to the form "bed to bench."

- World-class translational research facility
- Highly trained and skilled, clinically licensed experts
- ISO 9001:2015 Quality Management System certified



Bridge between DISCOVERY and development of therapies



Hematology and Oncology

Achieve a better understanding of the cellular and molecular bases of blood cells and cancer.

UNVEIL and DEVELOP new clinical therapies & treatments. Reach our experts through <u>actc@sidra.org</u>

Zainab Awada, Watfa Ahmed, Anjud Al-Mohannadi, Zayana Ali, Mariam Lotfy, Reem Mohammad, Massimino Miele, Sara Deola, Chiara Cugno, Aylene Marquez, Che-Ann Lachica, Dhanya Kizhakayil, Michelle Esblaca, Sheanna Marie, Juvilyn Gusi. erts rtified



Stem Cells

Move innovative stem cell methodologies into accelerated clinical applications.

Genomic Data Science Core

Genomic Data Science Core (GDSC) enables researchers and clinicians at Sidra Medicine and external collaborators to efficiently manage, analyze and interpret their data. GDSC developed a clinical-grade WGS bioinformatics pipeline for pathology department at Sidra Medicine. Moreover, GDSC co-developed, with QPHI's bioinformaticians, a pipeline to process the QPHI over 30 thousand samples from QPHI. GDSC develops analysis pipelines to QC, process and analyze a wide range of datasets like WGS, WES, and (sc)RNA-seq, and deliver the results to the end users in a short turnaround time. GDSC works closely with the HPC team at Sidra to provide the necessary infrastructure to allow for rapid processing of datasets as well as storing and archiving the results securely and safely for internal and external collaborators.

Flow Cytometry Core

The Flow Cytometry Core provides a multifaceted phenomics platform dedicated to establishing cellular, molecular, and functional phenotypes that complement genomics, transcriptomics, and clinical phenotypic analyses of patients. The core aims to complement in depth and breadth the panels of diagnostic and investigational assays proposed in Sidra Medicine and in Qatar as well as establishing international collaborations. The core mission is to provide the technical and intellectual frameworks for the realization



Mohammed Abuhaliqa, Abdul Rahman Salhab, Ashraf Saifullah.

Khadega Ibrahim, Giusy Gentilcore, Enas Dawoud.

of the second pillar of Sidra Research Personalized Medicine agenda: "Establishing an Advanced Diagnostics program". The core provides sperm DNA fragmentation in-house developed assay, no other lab in Qatar, currently outsourced or not offered and provides advanced serological test for SARS-CoV-2: Home-grown assay tests for 11 antigens (SARS-CoV2 + 6 other human CoV's), leading to MOPH approval as rapid antigenic/serology test, BAU validated, quantitative and qualitative assay.

Advanced Imaging Core

Aiming to empower precision medicine through advanced imaging technologies, the Advanced Imaging Core (AIC) specializes in enabling precision medicine by providing cuttingedge imaging technologies and expertise to researchers, clinicians, and innovators. Our mission is to accelerate discoveries and innovations that drive personalized healthcare solutions, with applications spanning oncology, genomics, drug development, and more. Our state-of-the-art facility is equipped to support a wide range of projects, from high-throughput screening to in-vivo imaging in zebrafish models. Through our collaborative and customerfocused approach, we aim to make advanced imaging accessible, intuitive, and impactful for every researcher and clinician we serve.

Clinical Trials Office

The Clinical Trials Office (CTO) serves as a centralized coordinating office for Clinical Research and Clinical Trials aiming at guiding researchers and clinicians through the local regulatory pathway (IRC, IRB, MoPH), and at conducting scientifically and ethically sound human subject research. Through the allocation of Research Coordinators' resources trained in the set-up and conduct of clinical research, the CTO's role and objective are to lead and oversee



Farhia Abdullahi Ali, Abbirami Sathappan.

Divya Rangarajan, Treesa Jilson, Blessing Dason, Ilyas Ali, Chinnu Samuel, Antonella, Cioce, Asma Jamil, Muhammad Ali Hameed, Dharati Tapse, Fathima Abubacker, Gulafsha Fatima, Mona Mahmoud, Aisha Kamil.

studies from protocol design to close-out. The CTO supports investigator-initiated and sponsored studies and functions as the point of contact for external sponsors including Contract Research Organizations (CROs) and Pharma companies. The CTO strives to enable both Academic and Pharma-sponsored Clinical Trials with benefits encompassing strategic value, public health, as well as financial value.

Research Operations Innovation and Strategy

The Research Operations Innovation and Strategy is led by Dr. Marla Beerens. Dr. Beerens pursued in Health law and obtained a Bachelor of Health Organization, Policy and Management and Economics at Maastricht University. She has a Master of Philosophy in Clinical Epidemiology and Health Technology Assessment and was awarded over 550.000 Euros in grant funding to conduct a multicenter randomized control trial, which helped her complete her PhD in Public Health. In the last 5 years at Sidra Medicine, Marla has focused on managing strategic, operational, and improvement programs while working in the Business Excellence Team.

Department Overview

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 Outcomes & Reporting Office looks after strategic reporting, communications, education, and outreach. The Laboratories & Biosafety Office ensures labs are in good working conditions and safe practices. The Grants Office manages external and internal competitive awards. The Business Office handles budgets, finance, and procurement matters.



Research Support Staff team has played a vital and multifaceted role in supporting the Research Branch since its inception in 2013. The diverse backgrounds in chemistry, psychology, literature, nursing, and business administration bring a broad range of expertise and perspectives to the team. This mix contributes to the team's success in managing both the administrative and organizational aspects of the research process.

Key Areas of Support Include:



Overseeing the organization of major research conferences and symposiums, with a track record of successfully managing about 12 such events over the last nine years. This demonstrates not only logistical and organizational skills but also the ability to coordinate large-scale projects in a fast-paced, academic environment.



Team Values



Zubair Pote, Maricris Salud, Dalia Salem, Dhanya Vasudeva, Rindai Magezah, Chetaram Nepali, Jeanna Marie, Patricia Hachem, Marla Beerens, Jason Ramm, Irem Mueed, Ashok Kumar, Mujubur Rahman, Farhan Mohammad, Roshan Ashraf, Mariam Yasser Nofal.

Raoudha Essaker, Nelly EL Mistekawy, Rana Hamada, Shweta Chaudhary, Paulina Mari Jalandoni.

38 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

Research Support

Handling committee meetings, preparing agendas and minutes, managing HR-related matters within the Research department, and overseeing logistics for travel and events.

> **Events and Conference** Management

The emphasis on core values like Care, Efficiency, Innovation, Teamwork, Transparency, and Trust reflects a commitment to maintaining high standards while also fostering a collaborative and supportive work environment. Discipline, punctuality, problem-solving, and confidentiality are also clearly integral to the team's operations, ensuring the smooth running of the department.

TECHNOLOGY TRANSFER AND INNOVATION

The Technology Transfer and Innovation function at Sidra Medicine plays a pivotal role in fostering innovation and translating groundbreaking research into tangible outcomes. The Technology Transfer function receives all invention disclosures from across the hospital for assessment and recommendation.

This milestone is a testament to our expertise and dedication to supporting innovators at Sidra Medicine. As part of its strategic efforts to support national innovation, the team is currently engaged in a study titled "Cultivating Innovation at Qatari Knowledge Institutions: A Case Study Approach". The study, led by Dr. Marla Beerens (Lead PI) and Mariam Nofal (Co-PI), is supported under the Pathway Towards Prosperity Fund by QRDI.

This initiative aims to develop policies and strategies that nurture innovation and IP creation within Qatari institutions, positioning Sidra Medicine as a leading hub for medical innovation. The Technology Transfer and Innovation function works closely with innovators across the institution, assessing their invention disclosures and recommending optimal IP protection strategies. Through collaborations with key stakeholders, such as the Legal Department, the team ensures effective management of Sidra Medicine's IP portfolio, safeguarding and advancing the institution's innovative outputs.

Intellectual Property (IP) encompasses a range of assets such as patents, trademarks, copyrights, and trade secrets that protect the tangible outcomes of innovation and creativity. In the context of research, IP is particularly critical as it secures the rights to novel discoveries, enabling innovators to retain ownership and control over their work.

This is essential for translating research into practical applications, such as diagnostic tools, therapeutic solutions, and implementation research projects. IP serves as a bridge between groundbreaking ideas and their realization in the marketplace. It acts as a trading tool that facilitates partnerships with companies and collaborators who possess the resources and expertise to advance innovations and bring them to market.

By protecting these assets, Sidra Medicine ensures that its research outputs can attract investment, foster collaborations, and ultimately benefit patients and the broader community.

Technology Transfer Process

What is Intellectual Property (IP) and Why is it Important?

TRADEMARKS

Beyond IP management, the function actively facilitates engagements and collaborations with industry partners, bridging the gap between research and commercialization. By connecting researchers with industry leaders, the team fosters opportunities for joint development, licensing, and innovation-driven partnerships, further advancing Sidra Medicine's mission of driving medical and scientific breakthroughs.

This year, the function received five new invention disclosures and successfully elected two patent applications for full patent filings, marking significant progress in our intellectual property portfolio.

If you have an invention, research proposal that encompasses a novel implementational idea, idea for IP protection, or even a publication draft you would like the team to review, submit your invention disclosure or queries to Mariam Nofal at mnofal@ sidra.org. The Technology Transfer and Innovation team is here to help bring your ideas to life.

CAPACITY BUILDING

Outreach and education at Sidra Medicine's 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 Medicine Research Branch 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 our research branch 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.

12 VISITING SCIENTISTS:

Mahmoud Mohamed, Hamad Medical Corporation Farook Al-Ajli, Al Ghannas Qatari Society Naziha Alem, University College London Ayesha Yasmin, Hamad Medical Corporation Tawa Olukade, Hamad Medical Corporation Arash Rafii, Weill Cornell Medicine Qatar Sawssan Ahmed, California State University Abeer Fadda, Heidelberg University Cristina Maccalli, Università degli Studi di Milano Salim Bougarn, François Rabelais University Nicholas Van Panhuys, University of Otago Mohamed Alaghbar, Sidra Medicine Alumni

69 Externs and volunteers enrolled in 2024

11 PhD students in 2024

25 Local and international universities

STUDENT TESTIMONIALS

Dena Al Dasooqi, MSc

After graduating with my Bachelor's degree in Nutrition and Dietetics in Australia, I was eager to gain hands-on experience in nutrition research. Joining Dr Annalisa Terrenegra's Precision Nutrition team as an extern was the perfect way for me to do that. During my time here, I gained invaluable knowledge and skills which have built my confidence as a young researcher. Among other tasks, I worked on raw data processing, data analysis, and manuscript writing, and I had the pleasure of seeing our project through to completion. To top it all off, I found the most welcoming and supportive colleagues, and I had the chance to learn from like-minded people and professionals in the industry. This experience has inspired me to continue growing in the field, and it will be a huge asset to my future studies and research career. Thank you to everyone who I crossed paths with, you have made this a unique and unforgettable experience.

Fatima Mohamad Ahmad, Ph.D

I am a Ph.D. candidate in Biological and Biomedical Sciences at Hamad Bin Khalifa University, conducting cutting-edge research in collaboration with Sidra Medicine. During my externship in Sidra, I honed expertise in advanced wet and dry lab techniques, including DNA methylation analysis, 16S rDNA sequencing, and computational biology. My research is centered on maternal and fetal health, with a specific focus on multi-omics approaches, precision nutrition, and the discovery of gut microbial and epigenetic biomarkers. In addition to my research, I have actively contributed to scientific writing, aiming to publish in esteemed peer-reviewed journals. My work has been recognized through multiple awards, including Best Poster and First/ Second Place awards at prestigious local and international conferences. These experiences have not only enhanced my technical and analytical skills but also fostered personal and professional growth, equipping me with a strong foundation to excel in my future career in biomedical research.

Abdallah Al Miski, High School IB Student

My experience at Sidra Research under the mentorship of Ms. Zenab was rewarding and wonderful. I appreciate the organization of the program, aligning with my school's schedule and being timely managed. At first, I was taught how different researchers operate, beginning with receiving samples, writing pedigree charts, looking through DNA sequences that cause mutations, utilizing AI in research, and using statistics to formulate conclusions. I really thank Sidra and Ms. Zenab for providing me with this valuable experience.

Maria Esteves, MSc

This year I had the privilege of being an extern in Dr. Avella's Reproductive Biology lab as part of my MSc in Human Embryology and Developmental Biology at the University of Aberdeen. This experience provided me with lifelong skills for the research industry and the workplace in general. While studying the genetic causes of infertility, I was able to learn and develop key lab skills such as immunostaining and confocal microscopy, which was incredibly valuable for my future as a scientist. Additionally, I was also able to strengthen my ability to present scientific research in professional settings and contribute towards ongoing research for publications. I truly relish my experience at Sidra and getting to work in a team and institute full of remarkable scientists who taught me so much.

Noura Aldous, PhD

Pursuing my PhD at Sidra Medicine under the mentorship of Dr. Essam Abdelalim has been a magnificent opportunity. His expertise in stem cell biology and commitment to groundbreaking research have truly been inspiring. Throughout my time here, Dr. Abdelalim has not only guided my scientific inquiry but has also fostered an environment of intellectual curiosity and growth on both personal and academic levels. His approach to research emphasizes innovation, collaboration, and a deep understanding of the intricacies of biomedical science. At Sidra, I have had access to world-class facilities and a vibrant research community that has enriched my academic journey. The knowledge and experience I have gained here is invaluable, shaping me into a confident and capable scientist. I am grateful for the mentorship, which has been instrumental in my personal and professional growth throughout my PhD journey.

Zahra Yusuf, MSc

I am currently pursuing a Master's degree at Hamad Bin Khalifa University (HBKU) under the supervision of Dr. Luis Saraiva. Over the past nine months in Laboratory of Disease Modeling and Therapeutics, I have significantly developed my skills in communication, teamwork, problem-solving, and hands-on research techniques. I have gained the ability to work independently within the fields of zebrafish research and molecular biology. Additionally, I had the privilege of presenting my research findings at PMFG 2024, an experience that further reinforced my commitment to the field. I am grateful to be part of Saraiva's lab, where I have had the opportunity to collaborate with talented colleagues and learn from their expertise. This experience has deepened my passion for research and motivated me to continue pursuing a career in this field.

Nafisa Fadulelmula, BSc

My externship in the Pluripotent Stem Cell Lab with Dr. Essam Abdelalim at Sidra Medicine has been a rewarding experience. I gained hands-on expertise in differentiating iPSCs into pancreatic beta cells and developed essential skills in aseptic techniques, q-PCR, western blot, and immunostaining. This work also improved my understanding of signaling pathways involved in pancreatic development. Working under Dr. Essam's guidance helped me strengthen my ability to analyze and interpret experimental results, improving my problem-solving and critical thinking skills. Collaborating with a skilled and supportive team allowed me to learn from others and develop a collaborative mindset. Presenting findings to colleagues also enhanced my communication and presentation abilities. This externship has not only strengthened my technical and analytical capabilities but also fueled my passion for stem cell biology and regenerative medicine, preparing me for advanced biological and biomedical research.

Jamie Navarro, High School IB Student

I did a two-day shadowing experience with Dr. Younes Mokrab's team at the Medical and Population Genomics Lab, and I can say with confidence that this experience was a definite eye-opener to how innovative and exciting the field of medical research can be. I gained in-depth knowledge and understanding of the incredibly detailed process of medical research; the wonderful experts were excellent in explaining the concepts of their respective work. Understanding each critical step when solving cases, I developed my research and critical thinking skills. Incorporating artificial intelligence to aid in modern-day research was especially innovative and interesting to see.

FRESH GRADUATES

0000

Mohammed Janahi University College London PhD in Medical Imaging

0000

Abbirami Sathappan University of Genova PhD in Haemato-Oncology and CTIM

0000

Parul Singh Hamad Bin Khalifa University PhD in Genomics and Precision medicine

0000

Randa AlYafie Hamad Bin Khalifa University PhD in Genomics and Precision Medicine

0000

Asmaa Al-Naama Hamad bin Khalifa University MSc in Genomics and Precision Medicine

0000

Eman AbuMoussa Staffordshire University MSc in Molecular Biology

Fajr Mansoor Al-Marzooqi Hamad bin Khalifa University MSc in Biological and Biomedical Sciences

0000 Shaikha Alabduljabar

Noora Al-Muhanadi Hamad bin Khalifa University MSc in Genomics and Precision Medicine

> 0000 Tariq Abu Saqri University of Liverpool MSc in Data Science and Artificial Intelligence

Mariam Nofal Universita degli Studi de Turin LLM Master of Laws in Intellectual Property

> 0000 **Maricris Salud** Ateneo Graduate School of Business Master's in Business Administration

Hamad bin Khalifa University MSc in Genomics and Precision Medicine

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Sidra Medicine Introduces Genomics to High School Students

Sidra Medicine Continues Partnership in Empower Generations Consortium

Sidra Medicine has taught more than 400 high school students in its Introduction to Genomics (InGen) student programme in 2024. Launched in October 2023 by Sidra Medicine's Genomic Core team and lead by Iman Al Azwani, InGen focuses on providing students access to real-life research environments and helping them in their future career choices in science and research.

In collaboration with the Ministry of Education and Higher Education (MoEHE), the programme has welcomed 11th and 12th grade students from 35 public and private schools across the country.Sidra Medicine has a dedicated Biosafety Level 1 Laboratory for the InGen programme, where students have a chance to learn both theoretical and practical components in genetics including DNA extraction, DNA quality checking and DNA amplification. It provides a safe and controlled environment where students can conduct real-world experiments and enhance their understanding of complex genetic processes.

The Empower Generations Consortium is an initiative founded by Qatar University to which Sidra Medicine has been a proud partner since its conception in 2019. Empower Generations Consortium solely aims to enhance the conceptual development of life sciences and grant youths life fulfilling careers that help them to dynamically engage with the societies, build their inner strengths, reshape their selfconfidence, heighten their competency levels, improve their interpersonal skills, initiate their social contribution, and influence their adeptness to the surroundings.

It is the only project-based consortium that tackles the career related challenges from the secondary education's platform and connects the bridges between the (schoolsuniversities- employments-national demands).

The project facilitates genuine engagement of high school students with professors and researchers, helps the new generations to meet their role models at various healthcare facilities, strengthens the national commitments and re-enforces responsibility.

This year, Sidra Medicine Research participated in a hands-on session led by Tala Abu Arja representing the Zebrafish Core Facility and the work done there. Students were introduced to the zebrafish model and the logic behind its application to human genetic research. The group of students were also invited to experience the larvae stages as seen through the microscope in a practical application.

Qatar Academy of Science and Technology Internship

Qatar Academy for Science and Technology (QAST) is a high school designed to develop Qatar's next generation of science, technology, engineering and mathematics (STEM) innovators. QAST students from across Qatar having a passion for science and technology and a desire to access the best universities and STEM related careers are supported and empowered to become innovators who master the knowledge and skills required to create solutions for challenges that face Qatar and the world.

For five Tuesdays in January and February, five 12th grade students from Qatar Academy of Science and Technology (QAST) joined an intense internship program at the Sidra Medicine Research facilitate as part of their mandatory "career buffet" school initiative. In the internship program, researchers identify parts of their research projects that can be supported by QAST grade 11 & 12 students, and provide technical training and mentorship for them. Sidra Medicine representatives start with an overview to QAST and present their ongoing research, explaining how the students can support.

This helps researchers and students to reflect and match their interests and competencies, which also adds value to student distribution among the different labs, as per their interests. QAST Internship is a 5 week program (total of 35 hours). The zebrafish facility works with researchers and clinicians leveraging the functional validation in the zebrafish model shedding light on gene discoveries, clinical presentations, disease prognosis, and treatment strategies.

The facility offers pediatric disease models for neurological disorders, cardiovascular diseases, congenital anomalies, and other diseases, making it the ideal visual learning platform for aspiring young scientists. The International School of London in Qatar has a mission to empower learners to lead through compassion, collaboration, creativity, and critical thinking. On March 24, students from the International School of London had a memorable experience visiting the Zebrafish facility.

52 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

International School of London Visit

Our staff guided them through zebrafish husbandry, genetic alteration, and behavioral studies, providing essential insights into biological research procedures.

Hands-on experiences with living organisms piqued their interest and enhanced their comprehension of scientific ideas, instilling a desire for scientific research. Immersed in the intriguing world of the zebrafish model, students saw cutting-edge research develop in real time. We appreciate our talented participants' enthusiasm to learn and explore alongside us.

Hamilton International School Visit

QRDI Corporate Innovation Leaders Program

On February 19, 2024, 11th-grade students from Hamilton International School in Doha embarked on an enlightening journey into the realm of precision medicine during their recent visit to Human Disease Modeling Laboratory led by Dr. Luis Saraiva at the Research Branch of Sidra Medicine.

Immersed in the cutting-edge world of medical research, the students delved into the intricate web of precision medicine, and learned how it is being implemented at Sidra Medicine. Guided by Dr. Saraiva and his team, they explored the innovative methodologies employed in disease modeling and gained insights into the transformative potential of personalized healthcare approaches. The immersive experience included guided tours of research labs, the zebrafish facility, and the hospital healing gardens, providing students with a firsthand glimpse into the evolving landscape of medical science.

This experience sparked their curiosity and nurtured a deeper appreciation for precision medicine's pivotal role in shaping the future of healthcare. In a globalised business landscape where technology is evolving at a rapid pace, corporates wishing to capitalize on the opportunities presented by this technological evolution have to adapt their strategies and be nimble. Adopting innovation allows corporations to gain a competitive advantage, enabling them to attract talent, bring innovative new products to market and further business objectives. To enable this, QRDI Council has launched a Corporate Innovation Leaders Program for large local enterprises. The program brings together strategy leaders from corporations that play a vital role in Qatar's innovation ecosystem.

The program aims to expand each organisation's internal capacity for carrying out innovation activities. Participating entities receive support in developing their own innovation strategies to boost technology adoption, and encourage partnerships with R&D-intensive businesses to deploy,

implement and co-develop innovative solutions that enhance competitiveness and performance. A team of delegates from Sidra Medicine, including from the research branch, attended the 6th edition of the prestigious QRDI Corporate Innovation Leaders Program in April. The program, delivered in partnership with Alchemist Accelerator, brought together multi-sector teams from around the Qatari innovation landscape for a week of inspiring talks, focused learning, mentorship and networking at Qatar Science and Technology Park.

The program culminated in the Sidra Medicine team pitching a very well-received idea to develop an online platform to transform continued care at Sidra Medicine for the benefit of patients in Qatar and the wider Arabic speaking world. Sidra Medicine delegates included Farhan Naim, Nour Saleh, Patricia Hachem, Maha Al-Henzab, Abdul-Aziz Zaghmout, and Aljori Al-Ajji.

INTERNAL RESEARCH FUND

Sidra Medicine invited research proposals for the Internal Research Fund 2024 (IRF 24) awarded in Q1 2024. The purpose of this intramural grants program is to support competitive and innovative research initiatives that improve the patient's outcome by leading to early detection, better prevention, improving diagnostics, and treatments. Special attention was given to disorders that are prevalent in Qatar and that affect Sidra Medicine patients, and that fall within the following programs:

- Neurological disorders (e.g., autism, epilepsy)
- Cancer (e.g., pediatric solid tumors, leukemia)
- Genetic/metabolic disorders (e.g., rare genetic diseases, childhood diabetes)
- Immune dysregulations (e.g., IBD, primary immune deficiencies)

Applications need to be translational and outcome-driven; where clinical outcome improvements would be possible to measure and enabled by Precisions Medicine approaches. The primary purpose of IRF 2024 is to provide funds for activities that will lead to the development and submission of a competitive research proposal to an external agency (e.g. QRDI or others). The application should demonstrate how the proposed activities will enhance the probability of reaching definitive outcomes in a targeted, externally funded grant. It is mandatory for each project to have one LPI from the Research and the Clinical Department.

10 applications out of 32 submitted proposals got awarded with a total awarded amount of QAR6,771,698.

Awarded

Successful proposals receiving funding

Submitted

Total proposals submitted for consideration

Lead PI	Co-LPI	Project Title
AL-MARAGHI, Aljazi	BEN OMRAN, Tawfeg	Genome First: Building a gold-stan- dard resource to accelerate newborn genomic screening in Qatar
AUGUSTINE, Tracy	YAJAMANYAM, Phani Kiran	Development of a Clinical Diagnostic Methodology for Early Identification of Non- IgE mediated Cow Milk Allergy and Prevention of FPIES Induction in pediat- ric subjects (NIGE-CMA Study)
FAWZI, Mahmoud	MOKRAB, Younes	Genomic medicine of neuromuscular disorders
RAYNAUD, Christophe	SALEH, Ayman	SLFN11 in Pediatric cancer
KARIM, Mohammed Yousef	LO, Bernice	High Frequency In-home Transcrip- tomics in Kids' Inflammatory Diseases (Hi-FIT KID) study
HENDRICKX, Wouter	OZER, Erdener	Advanced Pediatric Omics
AL KHODOR, Souhaila	RAPPAI, Shiga	Delineating the mechanistic role of gut microbiota associated Neuropathy in children with type 1 diabetes (T1D)
SYED, Najeeb	AL-SALEH, Rehab	Harnessing the potential of RNA-Seq as a complementary diagnostic method for WGS-negative Mendelian patients
THORNTON, Lisa	MOKRAB, Younes	Improving the diagnostic yield of cere- bral palsy
AHMED, Ikhlak BRANCO, Ricardo	ALYAFEI, Khalid VAN PANHUYS, Nicholas	Early identification and prognosis of pediatric sepsis and organ dysfunction phenotypes using RNA and circRNA expression

EXTERNAL GRANTS

23 total Grants from the Qatar Research Development and Innovation Council (QRDI) and other external funds were awarded to Sidra Medicine in the last 18 months. Total awarded amount is more than QAR 22M.

Breakdown of Grant Application In The Last 18 Months

Grant Application	Submitted	Awarded
Path Towards Precision Medicine (PPM 06, PPM 07)	25	3
Academic Research Grant (ARG 01, ARG 02)	39	5
Highschool Research Experience Program (HSREP05 HSREP06)	2	2
Conference and Workshop Sponsorship Program (CWSP23)	4	2
Pathways Toward Prosperity (PTP 01)	1	1
Economic Sustainability Call (ESC01)	1	-
Displaced Arab Researcher (DAR01)	2	TBC
Equine Research Call (QMRC01)	2	1
Open Innovation Call	4	4
Best Representative Image of an Outcome (BRIO08)	1	1
Technology Development Grant (TDG)	3	TBC
Leading House for the Middle East and North Africa "Research Partnership Grants"*	1	1
Orphan Disease Center, IQSEC2 Research and Advocacy Foundation Research Grant Program 2023*	1	1
G-Rex Grant Program Plan*	1	1
Global Grants for Gut Health call 2024*	1	1
Pfizer Independent Education Grants*	1	-
Orphan Disease Center, Million Dollar Bike Ride (ODC MDBR) Pilot Grant Program*	1	-
Incubator Grant 2024 Call from the Mito Foundation*	1	-
Alex Lemonade Foundation Crazy 8 Initiative*	1	-
Cure Epilepsy Grant Opportunities*	1	-
2024 CDKL5 Program of Excellence Pilot Grant Program*	1	-
Waterloo Foundation: Child Development*	3	-
Childhood Arthritis & Rheumatology Research Alliance CARRA*	1	-
Eagles Autism Foundation Grants: Pilot Grants*	1	-
Collaborative Pediatric Cancer Research Awards Program*	1	-

*International grants and awards

TOP ORIGINAL PUBLICATIONS

01

and Pulmonary Function Abnormalities at 3-year post COVID-19 Hospitalization: A Longitudinal Cohort Study. Authors: Osamah Alwalid. Journal: European Respiratory Journal.

02

Long-term Radiological The immunopathological landscape of human pre-TCR α deficiency: From rare to common variants. Authors: Andrea Guenoun, Taushif Khan, Manar Ata, Fatima Al Ali, Nico Marr. Journal: Science.

(0)4,

Integrated care of diabetes during pregnancy: a Qatari nationwide cohort. Authors: Ibrahim Ibrahim. Journal: EClinical Medicine.

05

Diverse Asian Genomes. Authors: Sonia Davila.

07

Predisposition footprints in the somatic genome of Wilms tumours. Authors: Gordan M Vujanic. Journal: Cancer Discovery.

Impaired development of memory B cells and antibody responses in humans and mice deficient in PD-1 signaling. Authors: Fatima Al Ali , Mahbuba Rahman , Nico Marr. Journal: Immunity.

10

Factors associated with successful liberation from continuous renal replacement therapy in children and young adults: analysis of the worldwide exploration of renal replacement outcomes collaborative in Kidney Disease Registry. Authors: Ahmad Kaddourah.

Journal: Intensive Care Medicine.

monocyte migration. Immunology.

03

Biallelic NAA60 variants with impaired n-terminal acetylation capacity cause autosomal recessive primary familial brain calcifications.

Authors: Sahar I Da'as, Khalid A Fakhro. Journal: Nature Communications.

A Catalogue of Structural Variation across Ancestrally

Journal: Nature Communications.

08

06

Major Adverse Kidney Events in Pediatric Continuous Kidney Replacement Therapy. Authors: Ahmad Kaddourah. Journal: Jama Network Open.

09

Trial of Selective Early Treatment of Patent Ductus Arteriosus with Ibuprofen.

Authors: Samir Gupta.

Journal: New England Journal Of Medicine.

11

Loss of the TRPM4 channel in humans causes immune dysregulation with defective

Authors: Satanay Hubrack , Christophe M Raynaud , Asha Elmi , Rafah Mackeh , Nourhen Agrebi , Hesham Al Saloos , Frank Schmidt, Bernice Lo, Amel Hassan. Journal: Journal of Allergy and Clinical

Covered Stent Correction for Sinus Venosus Atrial Septal Defects, an Emerging Alternative to Surgical Repair: Results of an International Registry.

Authors: Younes Boudjemline , Ziyad M Hijazi.

Journal: Circulation.

PUBLICATIONS ON JOURNAL COVERS

The book Alternative Splicing and Cancer co-edited by Dr. Ajaz Bhat explores the crucial role alternative splicing, a post-transcriptional process, plays in human health and diseases, particularly cancer. Diving deep into the complexities of gene expression and protein diversity, the book illuminates how abnormal splicing contributes to aggressive tumor formation, affecting cellular functions such as proliferation, survival, and immune evasion. With a focus on understanding molecular mechanisms, this book unravels potential diagnostic and prognostic targets, opening doors for enhanced anti-cancer treatment efficacy.

The cover picture of Diabetologia December 2024 issue shows an immunofluorescence micrograph of pancreatic islet organoids derived from normal induced pluripotent stem cells and iPSCs lacking regulatory factor X6, showing insulin, glucagon and nuclei. Aldous et al report that loss-of-function mutations in RFX6 do not impact the generation of iPSC-derived pancreatic progenitors. However, the absence of RFX6 leads to diminished islet cell formation and smaller islet organoids, which is associated with decreased expression of critical pancreatic endocrine genes and lower levels of the antioxidant catalase.

The paper titled Short-term consumption of highly processed diets varying in macronutrient content impair the sense of smell and brain metabolism in mice, co-authored by Melanie Makhlouf, Smija Kurian, Asma Al-Naama, Reem Hasnah, Neethu Venugopal, Diogo Manoel, and Luis R. Saraiva made the journal cover of Molecular Metabolism Journal, issue of January 2024. The image portrays artwork illustrating the link between the sense of small and the brain. Link to the article: <u>https://www.sciencedirect.com/</u> science/article/pii/S2212877823001710

Imaging Core at Sidra Medicine, has received Image of an Outcome in a QRDI-funded project. The award-winning image from the Pediatric Precision Oncology Initiative captures pivotal insights in pediatric cancer research, supported by contributions from Dr. Wouter Hendricks, Lead PI of the Pediatric Cancer Omics Laboratory, Dr. Erdener Advanced Imaging Core. This recognition reflects BRIO-8 Award celebrates Sidra Medicine's advanced imaging capabilities, reinforcing the institution's impact on pediatric oncology medicine for children in Qatar and the region.

GRANTS AND OUTPUTS 63

EVENTS AND ACHIEVEMENTS

64 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

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Precision Medicine and the Future of Genomics

PMFG 2024 Summit

Sidra Medicine marked a decade of innovation in genomic medicine with the opening of the 10th annual Precision Medicine and the Future of Genomics (PMFG 2024) Summit at QNCC. The three day event, which concludes 5 December welcomed 1100 attendees, featured over 50 distinguished speakers, showcased over 42 booths from sponsors and partners – highlighting the latest advancements in personalized healthcare, genomic research, and the ethical considerations surrounding this rapidly evolving field. Ms. Khadija Benganna from Al Jazeera Arabic was the Master of Ceremonies who opened the event with stories of hope featuring patient narratives.

PMFG 2024 Chairs, Dr. Ammira Al-Shabeeb Akil and Prof. Khalid Fakhro said: "PMFG serves as a vital platform for collaboration in precision health, bringing together the brightest minds to drive innovation and progress. Let's all work towards hope for future generations by building partnerships and pushing the boundaries of personalized healthcare." Sessions on Day 1 of PMFG 2024 focused on advancements in genomic newborn screening, rare disease diagnosis, and population genomic cohorts. Discussions explored the ethical, legal, and social implications of integrating genomic data into healthcare, strategies for early detection and personalized treatment, and the role of international collaborations in shaping global genomic research.

During her welcome remarks at the opening ceremony of the PMFG 2024 Summit, CEO Dr. Iyabo Tinubu-Karch said: "I am proud of the progress we have achieved and of how PMFG has evolved into a platform where the brightest talent and minds come together annually. Our shared insights into the latest research and best practices have a profound impact on patient populations globally, especially in the areas of genetic and rare diseases." Dr. Robert Green, Director of the Genomes2People Research Program at Harvard Medical School, delivered a compelling keynote address titled 'The Path to Lifelong Genomic Medicine': "Since the days of the Human Genome Project, people have envisioned a future where medicine transitions from reactive to proactive, and even to preventive.

We are entering a new era, with large-scale studies launching globally to generate evidence on how genomics can be used in screening. The next frontier is not just newborn sequencing, but lifelong genomics-informed healthcare that is accessible to all." Dr. Akil emphasized PMFG's key goals, stating: "Our mission is to promote innovation

in precision medicine, driving breakthroughs in genomics to enhance personalized healthcare and improve patient outcomes.

This summit also serves as a platform for empowering knowledge sharing, where insights on cutting-edge research, clinical applications, and emerging technologies in genomics can be exchanged to shape the future of healthcare." PMFG 2024 was held under the patronage of the Human Genome Organization (HUGO) and the Global Alliance for Genomics and Health, with Qatar Precision Health Institute as a strategic partner, the Children's Hospital of Philadelphia as a knowledge partner, Qatar National Bank as the Official Diamond Patronage and Sponsor, and Msheireb Museums as a community partner.

Sidra Medicine's First MENA Microbiome Conference

Our Research branch successfully organized the inaugural MENA Microbiome Conference, on September 28-29, bringing together over 800 attendees, including clinicians, scientists, and healthcare professionals from across the Middle East and North Africa (MENA) region. We had the opportunity to showcase our cutting-edge microbiome research in collaboration with clinical teams.

The two-day event, hosted in collaboration with the Ministry of Public Health (MOPH), Children's Hospital of Philadelphia (USA), Qatar Research Development and Innovation (QRDI), DH Arab, and Qatar Tourism Authority (QTA), featured a packed agenda of workshops, keynote presentations, and panel discussions. The main conference featured 38 expert speakers alongside presentations from 11 young scientists. Among the highlights was a panel discussion with prominent experts to establish with prominent experts to establish the MENA Microbiome Consortium. The conference underscored the growing potential of establishing the MENA Microbiome Consortium, leveraging local multidisciplinary expertise, and fostering collaboration with international centers. Ahead of the conference, a hands-on workshop was held on September 26th at our research facility.

The workshop focused on training attendees in microbiome sample processing and data analysis. It attracted 24 scientists from local institutions, such as Weill Cornell, Qatar Armed Forces, Qatar University, Primary Healthcare Corporation, and Hamad Medical Corporation. Additionally, international participants from Libya, Iraq, Tunisia, India and the UAE attended the workshop. The conference marked the start of promising new collaborations in microbiome research within the region.

Inaugural Conference on Primary Immune Dysregulation Disorders

Sidra Medicine, in collaboration with the MENAT-NIAID initiative, proudly hosted the Recent Advances in Primary Immunodysregulation Disorders (RAPID) conference in May, 2024. The inaugural event, which brought together close to 250 attendees, including global authorities in the field, experts from the Middle East, North Africa and Turkey (MENAT) and the National Institute of Allergy and Infectious Diseases (NIAID), demonstrated notable advancements in addressing the challenges and opportunities related to primary immune dysregulation disorders in the region.

RAPID also served as a powerful call to action for researchers, healthcare providers, and other stakeholders to help enhance the global understanding of these disorders.

The Career Development Panel session, featuring experts from Sidra Medicine, Hamad Medical Corporation, Qatar University, Weill-Cornell Medicine Qatar, Hamad Bin Khalifa University, and Mohammed Bin Rashid University (UAE), inspired the next generation of professionals to dedicate themselves to advancing care and research in this field.

The Jeffrey Modell Foundation's generous sponsorship at RAPID 2024, allowed young doctors and scientists from across the region, including those from lower-income countries, to attend and present their research. This investment in the future generation of researchers will have a lasting impact on the field of immune dysregulation.

Launch of Qatar's First Cord Blood Banking Service

Clinical Trials Offer Hope to Children with Rare Diseases

Sidra Medicine has launched a state-of-the-art cord blood banking service. This is the country's first local cord blood storage facility and will offer families a unique opportunity to preserve their newborns' stem cells in Qatar, for potential future medical needs.

Cord blood banking is the process of collecting and storing the blood from a newborn's umbilical cord and placenta after birth. The collection is painless, non-invasive, and risk-free for the mother and baby. Cord blood is rich in hematopoietic stem cells, which have the potential to treat various medical conditions, including certain cancers, blood disorders, and immune system diseases. Prof. Johnny Awwad, Chair of Women's Services at Sidra Medicine, said: "As the only facility in Qatar providing local cord blood storage, Sidra Medicine offers an unparalleled advantage by keeping the samples in the country." This ensures their optimal quality and immediate access when needed most, compared to having them shipped from abroad. All the cord blood samples will be sent to Sidra Medicine's Cord Blood Bank in its Good Manufacturing Practice (GMP) facility, where cryogenic freezing will allow them to be stored securely for over 30 years. Prof. Khalid Fakhro, Chief Research Officer at Sidra Medicine said: "Our cord blood banking service is another successful milestone for our precision medicine strategy."

Sidra Medicine aims to transform precision healthcare through clinical trials, offering a new lease of hope to young patients with rare and complex diseases. The Clinical Trials Programme at Sidra Medicine focuses on clinical trials that benefit children in the Arab region, particularly those with rare diseases that lack effective treatment. This initiative will position Sidra Medicine as a regional leader in clinical research, offering personalised treatment options to children with rare diseases.

The trials could offer children the chance to receive beneficial treatment whether it is a new drug, therapy, device or procedure — before it becomes widely available. Prof Khalid A. Fakhro, chief research officer at Sidra Medicine, said: "We are building towards undertaking complex research, including critical Phase 1 through Phase 4 studies, which will establish us as a regional leader in clinical trials. We were able to achieve this in collaboration with global academic and health partners,

Pharmaceutical companies and with the support of stakeholders such as Qatar Foundation (QF) and Qatar Precision Health Institute (QPHI). By becoming a regional leader in clinical trials, we will be able to bring innovative therapies to our patients, offering access to potentially life-changing outcomes to children with rare diseases." A historical lack of research within the Arab population has led to a significant data gap, limiting the development of effective and personalised treatments.

The clinical trials at Sidra Medicine will play a crucial role in addressing this gap, building a robust database of knowledge that will enable more effective and personalised treatment approaches tailored to the unique needs of the population. Sidra Medicine's clinical trials will be based on medicines, medical devices and medical procedures that have either been approved for adults but not yet studied for children or are not yet government approved.

Sidra Medicine and Msheireb Museums Launch Science Café

Largest Research on Rare Genetic Disease Risk in the Middle East

Sidra Medicine and Msheireb Museums launched their first Science Café series in 2024 with the concept based on inviting the public, including students and families, to connect with science and for scientists and engineers to informally share their research during a conversational exchange in a friendly setting. The Sidra Medicine-led Science Cafe focuses on groundbreaking advancements that are reshaping the country's healthcare delivery.

Through engaging discussions led by expert speakers, attendees discover how precision medicine can tailor treatment strategies for individual characteristics - from genetics to lifestyle. Dr Khalid Fakhro, chief research officer at Sidra Medicine, said: "Initiatives like the Science Cafe is our way to engage with the community and make science accessible to everyone as well as applicable to their day to day lives. Together, we can unlock the potential of precision medicine and its ability to transform lives and improve health for all." The Science Café series is free to attend and is open to everyone in the community who wants to learn about the latest technologies and applications in healthcare and research. Attendees will also be able to gain valuable insights that can empower them to make informed decisions about their health and well-being.

Dr Sahar Da'as, research manager at Sidra Medicine, who is leading the Science Café initiative, said: "We are really excited to start our own version of Science Café in Qatar and discussing science, research and health in bite sized pieces. This year, we will cover a range of topics and how precision medicine can help with diseases like diabetes, obesity, autism, paediatric cancers and address mental health to pregnancy and fertility challenges." A study on rare genetic diseases in a wideranging and diverse Middle Eastern cohort from Qatar conducted by our research team, has been published in the prestigious Genome Medicine journal. Titled Burden of Mendelian Disorders in a Middle Eastern Biobank, the study provides key insights into the distribution of risk for genetic diseases in Qatar, which will help with public health planning for the country's population and that of the wider region.

Led by Dr Khalid Fakhro and Dr Younes Mokrab and in collaboration with the Qatar Genome Programme (QGP), Qatar Biobank (QBB), and Hamad Medical Corporation (HMC) the study analyzed more than 6,000 whole genomes and corresponding health data from Qatar. The study established a comprehensive catalogue of disease mutations stratified by genetic ancestries, including the major groups of Peninsular Arabs, General Arabs, Persian Arabs and African Arabs; making it the largest such collection of genomes and clinical data to be investigated in a Middle Eastern population to date.

The researchers identified several damaging mutations specific to certain ancestries, known as founder mutations, that can serve as an invaluable epidemiological resource for policy making in the healthcare system. The information will also enable the healthcare system to offer screening and genetic counselling to families who may be at risk of inherited diseases.

They also identified novel disease-related genes/variants linked to common diseases in Qatar such as diabetes. By establishing a catalogue of Mendelian genetic variants, the study can also help improve annotation of global public genetics databases (ex., ClinVar database), especially for genetic variants that are common in the Qatari population and underrepresented in other databases. Ultimately, baseline genome studies such as the one by Sidra Medicine can help healthcare systems allocate resources more efficiently by targeting interventions to those patients who are most likely to benefit based on their genetic profile.

First Arab Middle Eastern Genetics Research in Autism Goes Global

Sidra Medicine celebrated the impact of its longstanding initiative, the BARAKA-Qatar study (Building a Resource for the Advancement of Knowledge of Autism in Qatar) on World Autism Awareness Day, marked on April 2 annually. The study, which has been at the forefront of Autism Spectrum Disorder (ASD) research in the region for several years and published its pilot phase in the prestigious Genome Medicine journal, has not only enhanced the understanding of ASD within the Arab world but has also pushed Middle Eastern genetics onto the global stage. Despite its global prevalence, the comprehension of ASD's genetic origins in the Arab world is historically limited.

The BARAKA study, conceived and conducted by Sidra Medicine in Qatar, shattered these boundaries, focusing on Middle Eastern genetics and their implications for ASD. Conducted in academic collaboration with the Hospital for Sick Children (SickKids) in Toronto, Canada, the study renders Middle Eastern genetics accessible and comprehensible on an international scale. This was made possible through SickKids's partnership with the Autism Speaks' MSSNG Whole Genome Sequencing Project (MSSNG), an innovative open-science based collaboration platform between Autism Speaks, Google, and the research community.

Dr Fakhro also noted that around 350 families, totalling approximately 1,500 participants, have been enroled from Qatar. He added: "Our collaboration with SickKids and MSSNG have assisted us in designing the BARAKA study to international standards, using shared models and analysis pipelines across our respective groups. Over the years, we have ensured that a number of trainees and sub-projects benefited from this joint mentorship. We continue to increase the size of our cohort to capture more individuals living with ASD in Qatar, which will aid in understanding underlying causes and development of personalised interventions in the future."

Sidra Medicine and Genialis Partnership: Enhancing Research and Innovation

Sidra Medicine is pleased to announce a collaboration with Genialis, an RNA-biomarker company. This partnership marks a significant step in our mission to advance precision medicine through robust research and innovative industrial collaborations. The integration of Genialis' advanced AI models into our research infrastructure allows us to delve deeper into the molecular diversity of our patient population. This collaboration ensures that our diagnostic and treatment protocols are informed by real-world data, making them more accurate and clinically relevant. It empowers us to predict patient responses to therapies more precisely, improving outcomes and setting new standards in pediatric cancer care. Dr. Wouter Hendrickx, Lead Principal Investigator of the Precision Oncology Initiative at Sidra Medicine, underscores the importance of this partnership: "Collaborating with Genialis represents a transformative step in our oncology program. By leveraging their technology, we can tailor treatments to the unique genetic profiles of our patients, enhancing the effectiveness of our therapies."

The partnership with Genialis enriches our research by incorporating diverse patient data into their AI models. This real-world data is vital for validating biomarkers and refining our understanding of disease mechanisms. It ensures that our research is not only cuttingedge but also directly applicable to the diverse populations we serve. Prof. Khalid Fakhro, Chief Research Officer at Sidra Medicine, highlights the broader impact of industry partnerships: Collaborations with the private sector are essential for accelerating research and improving patient care. Genialis, in particular, has built a remarkable record in developing clinically actionable RNA-based biomarkers. By integrating Genialis' suite of predictive algorithms into our workflows, we can begin to understand disease better and deliver personalized medicine.

These partnerships enable us to stay at the forefront of medical innovation, bringing the latest scientific advancements to our patients.

At Sidra Medicine, we believe that partnerships with industry leaders are key to driving medical innovation and improving healthcare outcomes. Our collaboration with Genialis is a testament to our commitment to research excellence and precision medicine. We are excited about the future advancements this partnership will bring and remain dedicated to enhancing the lives of our patients through pioneering research and innovative collaborations.

Distinguished Panelist at QPHI Launch

Sidra Medicine Article 'Up front' in Diabetologia

Chairperson of Qatar Foundation HH Sheikha Moza bint Nasser formally launched Qatar Precision Health Institute (QPHI) in April 2024, an umbrella institute bringing together existing strengths in the field of health sciences and genomics within Qatar Biobank and Qatar Genome Program to help accelerate the countrys adoption of personalized healthcare.

Dr. Kholoud Al-Shafai, a Staff Scientist - Associate Level at the Research Department of Sidra Medicine participated as a distinguished panelist at the event held at Qatar National Convention Center. Dr. Kholoud brought her expertise and insightful perspectives to the discussion on Precision Health. She discussed the genetic projects she currently leads and the implementation plans toward precision medicine. Dr Kholoud provided a clear vision for the role of Precision Health in transforming Qatar's healthcare landscape and highlighted on the necessity of intensive collaboration between various stakeholders, including researchers, healthcare providers, policymakers, and the community toward achiveing this goal.

Each month the Editor of Diabetologia selects five papers from the issue that merit special attention. These five papers are featured at the front of the print issue ('Up front') and appear on the Diabetologia homepage with a short piece of text summarising their importance.

Dr. Essam Abdelalim's article has been chosen to be featured 'Up front' in the December 2024 issue. The article titled *Deletion of RFX6 impairs iPSC-derived islet organoid development and survival, with no impact on PDX1+/NKX6.1+ progenitors,* PMID 39080045 explores how mutations in the RFX6 gene lead to monogenic diabetes with a hypoplastic pancreas. In this issue, Aldous et al investigate the mechanisms behind the impairment of pancreatic islet development caused by RFX6 loss-of-function mutations.

The authors utilise human embryonic stem cells and a gene-edited induced pluripotent stem cell model to investigate the role of RFX6 during differentiation into human pancreatic islets.

Findings suggest that RFX6 mutations primarily disrupt islet cell formation through increased cell death and suppression of essential pancreatic endocrine genes. The authors conclude that these insights may pave the way for potential therapeutic strategies, such as enhancing catalase levels, to mitigate diabetes related to RFX6 defects.

Zebrafish Leads the Way in Diagnosis, Treatment of Rare Paediatric Diseases

Study a Major Advancement in Cancer Microbiome Field

The Zebrafish Functional Genomics Facility at Sidra Medicine is leading the way in determining the cause and treatment of inherited disorders in children. The facility has adopted a remarkable approach to swiftly identify the causes of rare diseases and explore potential treatment solutions.

According to the Centre for Arab Genomic Studies, there are 2.8 million people living with rare diseases in the Middle East. Recognising the prevalence of inherited blood disorders like sickle cell disease and rare conditions in the Arab world, often stemming from single gene mutations, the hospital employs cuttingedge techniques to accelerate the diagnostic process. Specifically designed to address the unique challenges of diagnosing and treating rare diseases, Sidra Medicine focuses on paediatric patients, aiming to become a regional hub for this critical aspect of healthcare. Despite challenges posed by the diagnostic odyssey, typically taking five to seven years, Sidra Medicine has streamlined the process. Utilising the zebrafish, a species sharing 70 per cent of its genes with humans, researchers can rapidly study the effects of gene mutations. If the patient is found to carry a particularly unusual mutation, one that scientists have never encountered before, they begin the process of attempting to replicate the biology in a zebrafish embryo. Zebrafish offer advantages over traditional models, as they can be easily maintained in large numbers and exhibit rapid development, providing valuable insights within a week, equivalent to months in humans.

Sidra Medicine's approach allows for a quick transition from identifying rare diseases to potential treatments, with the goal of delivering personalised therapies within three months, a remarkable feat compared to the typical fiveyear timeline for diagnosis reported by the European Commission. Sidra Medicine also plans to collaborate with pharmaceutical companies for clinical trials, aiming to develop novel medicines tailored to patients with rare disease. Sidra Medicine has announced that one of its research studies published in Nature Medicine in May 2023 has been featured in the Year in Review section of Nature Reviews Gastroenterology and Hepatology, which focuses on key advances in the field of cancer microbiome in 2023. Titled 'Microbiota-based biomarkers and therapeutics for cancer management', the article provides a comprehensive overview of the latest discoveries and applications of microbiota-based approaches for improving cancer prevention, prognostication and therapy, with a focus on colorectal cancer. The research study from Sidra Medicine 'An integrated tumor, immune and microbiome atlas of colon cancer' by Roelands et al, revealed the impact of our immune system and microbiota on our ability to survive colon cancer. The study was done in collaboration with Leiden University Medical Center (LUMC) in the Netherlands. The study comprehensively characterised the genomic, transcriptomic,

immunological and microbial features of a cohort of 348 patients with colorectal cancer, sequenced at Sidra Medicine, with long-term follow-up data to devise a composite score for prognostication. The Sidra Medicine and LUMC team analysed various aspects of primary colon cancers, including the characteristics of cancer cells, cancer immune responses and microbiota composition.

They found that a specific composition of bacteria in the tumour and intratumoural immune response were associated with a more favourable prognosis for colon cancer patients. The Sidra Medicine study highlighted how combining a tumour inflammation score with the microbial signature into a composite score termed mICRoScore, maximised the prognostic performance, in which a mICRoScore-high group corresponded to a subgroup of patients with colorectal cancer with a 97 per cent and five-year overall survival rate.

Study Reveals Impact of Highly Processed Foods on Brain Metabolism, Sense Of Smell

Qatar Foundation Experts Discuss Biomedical, Ethical Perspectives on Genetic Modification

Researchers at Sidra Medicine in collaboration with teams from the Universidade Federal do Rio Grande do Sul (Brazil) and Monell Chemical Senses Center (USA), have published a groundbreaking study in the journal Molecular Metabolism, demonstrating how the short-term consumption of highly processed diets can impair brain metabolism and the sense of smell (olfactory) functions in mice.

Led by Dr. Luis R. Saraiva, Principal Investigator and A/Director of the Human Disease Modeling and Therapeutics Division at Sidra Medicine, the research team undertook the study to explore the short-term impact of consuming highly processed foods, especially in light of increasing global obesity rates and health concerns. The study involved an elaborate experimental design where mice, over a period of less than two months were fed three distinct diets: a standard grain-based chow diet, a highly processed control diet, and a highly processed high-fat diet.

Researchers conducted a comprehensive series of tests, including behavioral and metabolic phenotyping, electro-olfactogram recordings, brain glucose metabolism imaging, and mitochondrial respirometry. Advanced (ribonucleic acid) RNA-sequencing was performed on samples from the nose and various brain regions to understand the genetic impact. The study, co-funded by Sidra Medicine and a grant from Qatar National Research Fund, adds to the existing literature on the health impacts of processed foods and raises essential questions about dietary choices in contemporary society.

A progressive move taken by Qatar to do generic screening for potential spouses has made it possible to identify hereditary diseases that a child may be born with but also take early intervention measures to prevent them. Dr. Sahar Da'as, Laboratory Manager of the Zebrafish Facility at Sidra Medicine, member of Qatar Foundation (QF), explains that if one or both parents carry a genetic mutation that increases the likelihood of their child inheriting a hereditary disease. However early intervention and planning can be done by screening that particular gene in the embryos' DNA to prevent its inheritance or alleviate the resulting symptoms at early stages. Pre-marital medical tests, which are mandatory in Qatar, include performing genetic testing for potential spouses to detect potential genetic

boundaries cannot be ignored. While some view genetic testing as a means to reduce the spread of hereditary diseases and improve the health of future generations, others believe that this approach may lead to ethical challenges such as (genetic) discrimination or pressuring individuals to make decisions that may not align with their personal values.

According to Dr. Mohammed Ghaly, Professor of Islam and Biomedical Ethics at QF's Hamad Bin Khalifa University (HBKU), Research Center for Islamic Legislation and Ethics (CILE), religious scholars encourage genetic testing in the premarital stage, so individuals are aware of potential health conditions their children may have and can make an informed decision. Dr. Ghaly underscores the considerable ethical debates surrounding genetic modification or gene editing, particularly in the context of germline gene editing. This form of modification entails the permanent editing of genes in foundational cells, rendering the reversal of changes challenging, if not entirely impossible.

Pioneering Clinical Whole Genome Sequencing

Award for the Global Grants for Gut Health

We are pleased to announce the successful implementation of our Whole Genome Sequencing (WGS) service, as an advanced diagnostic-grade genetic test for our patients. The successful implementation of WGS is the culmination of years of Research infrastructure development, expertise building, and steady collaboration within various divisions, including genetic pathology, quality, the clinical genomic laboratory, and genomic data science core.

The method has the power to transform pediatric healthcare at our hospital by enabling more personalized diagnoses with accuracy and speed. WGS is a comprehensive genetic test that analyzes an individual's entire genome to identify genetic variations linked to diseases. It can quickly detect almost any genetic issue, including rare conditions that might otherwise go undiagnosed. The availability of the test in Qatar, significantly eliminates the need to send samples abroad, reducing turnaround times from months to weeks. This is particularly crucial for critically ill patients where timely diagnosis can be lifesaving. WGS is a complex project that required a significant investment in advanced DNA sequencing technologies and coordinated teamwork.

The implementation was supported by the Illumina Corporation. As the cost of WGS continues to decrease, we will consider making it a routine part of its pediatric care services especially for young patients with unexplained health conditions. The program will also enable proactive and preventive measures for the hospital's patients including fetuses and young babies, by identifying genetic risks in advance and developing personalized. Dr. Souhaila Al Khodor, Director - Perinatal and Reproductive Health Division, and her team from the Laboratory of Microbiome and Biomarkers Discovery received a Nature award for "The Global Grants for Gut Health", thus winning the "Best Abstract Prize" for Translational Science. Titled: Multiomics Profiling for a Comprehensive Understanding of Immune and Microbial Signatures In Pediatric Inflammatory Bowel Disease, the study is funded by QNRF under NPRP10- 0125-170242 and is a collaboration between Dr. Al Khodor's research team and Dr. Elawad's clinical team.

The Global Grants for Gut Health (GGGH) is a competitive programme for investigatorinitiated research into the human gut microbiota, supported by Yakult and Nature Portfolio. As part of the International Human Microbiome conference in June 2024, the GGGH gave awards to 3 top abstracts submitted to the conference in different research categories, including translational science.

Sidra Medicine's project won the Best Abstract prize for translational Science from almost 600 abstracts that were evaluated. The patients were all recruited from Sidra Medicine, and the study started in 2018.

This study, which is highly aligned with Sidra Medicine's mission to advance clinical care for women and Children living in Qatar, recruited IBD patients seeking treatment at Sidra Medicine. Several biological samples and extensive data was collected from the patients.

Using the State-of- the-art facility in the research department at Sidra Medicine, a unique gene expression and microbial signature for each IBD subtype was identified, with clear distinctions during disease flare compared to remission and healthy controls. This comprehensive understanding of the microbial-immune system interaction has the potential to reveal novel mechanisms and, ultimately, facilitate the discovery of innovative therapeutic targets to treat IBD.

Award-Winning Study on Treatable Inherited Metabolic Disorders in Qatar

Pediatric Oncology Report 2023

Sidra Medicine published a groundbreaking study in the journal 'Genetics in Medicine, an official journal of the American College of Medical Genetics and Genomics (ACMG)'. The research titled "Mapping the Genetic Landscape of Treatable Inherited Metabolic Disorders in a large Middle Eastern Biobank" was the recipient of the journal's Editor's Choice Award.

The study was also awarded first place for 'abstract presentation' at the MENA Congress for Rare Diseases 2024 in Abu Dhabi. Inherited Metabolic Disorders (IMDs) are a group of rare conditions caused by enzyme deficiencies with a genetic basis that disrupt the body's metabolism. Although each IMD is rare, their collective incidence is considerable, commonly estimated at 1 in 2,500 to 5,000 live births. Led by Prof. Khalid Fakhro in collaboration with (Qatar Precision Health Institute) QPHI, the study analyzed the genome sequencing data of more than 14,000 participants from Qatar Biobank focusing on 125 genes associated with 115 treatable IMDs. The study revealed a significant carrier frequency and disease burden for 46 treatable IMDs within the Qatari population. The results showed that approximately 1 in 13 unrelated individuals (eight percent) were carriers of at least one of these IMDs, with thirty-one participants (0.2 percent) carrying more than one disease-causing variant. Dr. Geethanjali Devadoss Gandhi, Senior Postdoctoral Fellow at Sidra Medicine and the first author of the published article said: "Our findings underscore the immediate need for comprehensive research and its potential to reshape public health policies, such as carrier and newborn screening programs, to address this previously unrecognized health concern."

The study's findings offer valuable insights into the prevalence and complexity of treatable inherited metabolic disorders (IMDs) within the Qatari population, underscoring the need for further research and treatment options. To mark World Cancer Research Day and Childhood Cancer Awareness Month, Sidra Medicine, a member of Qatar Foundation, has published its 'Pediatric Oncology Report 2023.' This year's report presents information on the treatment modalities employed at Sidra Medicine and the remarkable outcomes achieved for its young patients. It also takes a deeper dive by exploring the specifics of leukemia and brain tumors, going beyond the previous reporting of solid and non-solid cancers statistics.

The report offers an analysis of pediatric cancer types treated at the hospital, with a clear distinction between solid cancers (69 percent) and non-solid cancers (31 percent). Regarding patient demographics, the majority are of Arab descent (60 percent), followed by Asian patients (32 percent). The remaining less than 10 percent represent other ethnicities.

Among the diagnoses, Leukemia emerges as the most frequent at 31 percent, closely followed by Central Nervous System malignancies at 23 percent. Since launching its pediatric precision oncology initiative in 2018, treatment abroad has gone down from 20 percent in 2019 to only 11 percent in 2023.

Sidra Medicine's commitment to pediatric cancer care is highlighted by the upcoming launch of Qatar's first pediatric Hematopoietic Stem Cell Transplant (HSCT) program. This initiative, in partnership with Children's Hospital of Philadelphia, will provide life-saving treatment for children with cancer by replacing diseased bone marrow cells with their own healthy stem cells, or that of a donor.

Establishment of Institutional Animal Care and Use Committee (IACUC)

Sidra Medicine Advocates for Inclusive Genomic Databases

Our Research Department is excited to announce that we have now established our own IACUC committee for all animal research conducted in our organization. This committee will continue to be instrumental in ensuring that all animal-related research conducted within our institution adheres to ethical standards, regulatory guidelines, and institutional policies.

Its responsibilities include reviewing and approving research protocols involving animals, monitoring ongoing projects, and safeguarding the wellbeing of the animals involved. Comprised of interdisciplinary experts in fields such as veterinary science, research ethics, and animal welfare, the IACUC encourages close collaboration with researchers and staff engaged in animal research to uphold the highest standards of care and ethical conduct.

By fostering a culture that prioritizes animal welfare and promoting responsible scientific inquiry, the committee aims to make a positive impact within our organization's research community and beyond. We look forward to the positive impact that the IACUC will have on our organization's research community and the broader scientific community. In an article titled Addressing Inequity in Medical Research: The Need to Expand Genomic Databases published on Biopharma Trend, Prof. Khalid Fakhro addresses the critical need to address inequity in medical research by expanding genomic databases including Qatar and the region has been emphasized by the Chief Research Officer at Sidra Medicine, Prof. Khalid Fakhro. He shed light on the importance of global representation in databases to make breakthrough discoveries in health conditions and treatments. Prof. Fakhro acknowledges the progress made but emphasizes the long road ahead in making genome databases representative of global populations.

The underrepresentation of certain populations poses a significant challenge to the development of equitable and effective healthcare solutions. Prof. Fakhro points out that the lack of diversity in genomic databases can result in less effective care, as treatment approaches may vary based on genetic makeup.

The Middle East, with a population exceeding 400 million people, remains among the most underrepresented in genomic databases. Prof. Fakhro stresses the need for local pediatric databases to address the unique healthcare needs of children. The lack of diversity not only contributes to healthcare inequality but also limits medical research in the region.

To overcome the challenges, Prof. Fakhro emphasizes the importance of government and academic support, along with industry partnerships. He calls for increased international pharma and tech presence in the Middle East, citing the growing global next-generation sequencing (NGS) informatics market, expected to reach \$4.3bn by 2030. Prof. Fakhro anticipates that groundbreaking research and treatment development will benefit research bodies, industry partners, and local populations alike, paving the way for a more inclusive and equitable future in precision medicine.

International Day of Women and Girls in Science

Launch of IT Council

Sidra Medicine celebrated a very happy International Day of Women and Girls in Science with our female colleagues at Sidra Medicine. CEO Dr. Iyabo Tinubu-Karch said: "Whether it is diagnosing a health condition; finding a cure or a pathway to treat a patient; developing an IT or engineering platform to enable smoother processes - women have played a pivotal role across science, research, pathology, technology and engineering at Sidra Medicine.

Their excellence in these fields speaks of their passion in pursuing diverse careers. Thank you for your hard work and positioning us as an organization committed to offering personalized healthcare for all our patients and their families.

May you continue to be an inspiration for young women and girls everywhere." Women make important contributions to the field of research and innovation.

Whether they make fundamental scientific discoveries, find new ways to translate research results into real-world applications, or work to create optimal conditions for researchers to perform at their best, their impact is undeniable. However, women's contributions to research and innovation are often a lot less visible than men's. In addition, not only the research profession is stereotypically associated with men, but women remain widely underrepresented in science.

The Research IT Council, chaired by Dr. Sonia Davila, has been launched with the primary objective to streamline processes including gathering, evaluating, and prioritizing user requirements. The Council would serve as a centralized body to ensure that all requests are thoroughly reviewed, duplication is merged or eliminated, and only the most impactful and feasible functionalities are pursued.

This approach will not only enhance efficiency but will also ensure that projects remain aligned with end-users' needs and the division's strategic goals. The council meets monthly to discuss submitted requests. Kindly direct any questions or feedback on IMT matters to your PI or Core manager via email. They will ensure this is forwarded to one of the Team Members for inclusion in the agenda of the monthly meeting.

88 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

Goals:

- Aid IMT team in approving projects to pursue
- Help IMT team prioritize projects, both internally and with external dependencies
- Help allocate budget
- Discuss concerns, both for IMT and Research users. End-users could notify their PIs of any item they would like to be brought up to the Council.
- Drive alignment to CRO Goals

Team members:

Sonia Davila (Chair); Younes Mokrab (Core Member); Wouter Hendrickx (Core Member); Abdul Rahman Salhab (Core Member); Sasirekha Palaniswamy (Core Member); Farhan Mohammad Naim (Core Member); Ahmed Abdellatief (IMT Member); Ahmad El Khouly (IMT Member); Tarig Abu Saqri (IMT Member); Mehshad Hamza (IMT Member).

90 SIDRA MEDICINE RESEARCH | ANNUAL REPORT 2024

GENETICS

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120 27

109

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