Unlocking the Secrets of Craniofacial Development:
A Multidisciplinary Team Approach

How does our face take shape? It’s a question that may seem simple on the surface but hides layers of complexity beneath. Dr. Heather Szabo-Rogers, an Assistant Professor of Anatomy, Physiology, and Pharmacology at USask has embarked on a mission to unravel this enigma, and her approach involves a multidisciplinary team that could change the lives of countless individuals.

Major birth defects affect approximately 3% of children born in Canada each year. Among these, congenital craniofacial and cardiovascular anomalies stand out as the most common anomalies identified at birth, carrying profound impacts throughout a patient’s life. Birth defects often arise in both a patient’s heart as well as skull and soft tissues of the head because these structures develop from a shared population of precursor cells, using similar networks of genes. Disruptions in head and heart development can be attributed to genetic variants and exposure to environmental toxins, such as alcohol, nicotine, and air pollutants, during critical stages of in utero development.

Dr. Szabo-Rogers’ research is driven by a fundamental question: what are the genetic, molecular, physical, and genetic factors that contribute to congenital craniofacial anomalies? Specifically, her team is focused on understanding the prevalence of these anomalies in Saskatchewan. By unraveling the genetic variants associated with these conditions, they aim to determine their significance and role in malformation.

To tackle these complex questions, Heather and her dedicated team have intertwined two distinct approaches. Heather’s laboratory employs animal models, providing a controlled environment to explore the intricate processes of craniofacial development. Heather is forging new partnerships with Dr. Bita Hashemi, Assistant Professor of Pediatric Medical Genetics at USask and a medical geneticist for the Saskatchewan Health Authority. Dr. Hashemi, in her role as a medical geneticist performs genetic testing for patients who have been referred to her at the SHA Pediatric Genetic Program located in Royal University Hospital and Jim Pattison Children’s Hospital. This combination of both animal and human approaches accelerates the discovery of novel causes of birth defects tailored to individual patients.

Continued on pg 2…
Heather and Bita’s collaborative study, “A personalized medicine team for craniofacial and cardiovascular structural birth defects in Saskatchewan” seeks to determine the prevalence of congenital craniofacial and cardiovascular anomalies in Saskatchewan's children and youth. They are conducting a retrospective case review of patients referred to the genetics clinic with cardiovascular and craniofacial anomalies. This investigation will not only shed light on the scope of the issue but also identify variants for research studies aimed at understanding the molecular mechanisms of these diseases and potentially discovering treatments.

The impact of this research extends to multiple facets of our society. First and foremost, this team’s work has the potential to transform the lives of individual patients. By explaining the function of specific genetic variants, this research can help patients understand the potential risks to their children or siblings. Moreover, it may lead to the identification of actionable pathways or variants that could significantly improve patients' health outcomes. An excellent example of this is Heather’s work on Robinow Syndrome, where her team identified an upregulated signaling pathway and found an approved inhibitor that could potentially spare patients from surgeries.

Beyond individual patients, this research has implications for various organizations, including universities, government agencies, and medical colleges. The retrospective analysis of patients referred for craniofacial and cardiovascular anomalies over the past decade will provide critical data. By estimating the number of individuals affected by specific disorders, organizations can better allocate resources and plan interventions to support affected individuals effectively.

In conclusion, this research in craniofacial development is more than just a scientific endeavor; it’s a pursuit that has the potential to change lives, inform organizational strategies, and benefit society as a whole. As they delve into the intricate mechanisms shaping our faces, they bring us one step closer to unlocking the secrets of craniofacial development and improving the lives of countless individuals affected by congenital craniofacial anomalies.

Thank you to Dr. Heather Szabo-Rogers and Dr. Bita Hashemi for providing the content for this article. Dr. Szabo-Rogers would also like to acknowledge the following team members: Drs. Brian Eames, Julia Boughner, Dave Cooper, and Michele Collins.

This research is funded by: Saskatchewan Health Research Foundation 2023-24 Align Grant, Child and Youth Health, and Dr. Szabo-Rogers’ College of Medicine Start-up funds.

**Funding Opportunities**

**2024 Dean's Project Proposal Call Out**

Deadline for submissions is 4:00pm on November 9th, 2023!

For more information follow this [link](#)
Dr. Astrid Lang, Pediatric Resident, presented her research at the European Society for Paediatric Gastroenterology Hepatology and Nutrition (ESPGHAN) in Vienna, Austria in May! Her study was titled, “Effects of delayed/missed biologic infusion on children from Saskatchewan with inflammatory bowel disease during the COVID-19 pandemic” co-authored by Dr. Simone Nicol.

Congratulations to Pediatric Resident, Dr. Alyssa Zucchet for her poster presentation at the annual Canadian Pediatric Society conference in Halifax last week. Her presentation was titled, Reducing the risk of ATV-related Harm in Saskatchewan Children. Co-authored by Dr. Karen Leis.
Department of Pediatrics Research Report

Congrats to **Dr. Alan Rosenberg**, Department of Pediatric Faculty, being awarded a Canadian Rheumatology Association CIORA grant for the study, “Identifying Facilitators of and Barriers to Digital Health Literacy in Pediatric Rheumatic Disease Patients and Their Families”

A Rosenberg, M Jariwala, T Kerr, R Sinha, J Bajwa, D Goodridge, J Stinson, Craig Eling, E Linsley, H Wiebe, B Zerr

Learn more about it CIORA [here](#)

Congratulations to **Dr. Alan Rosenberg** who was awarded a Canadian Academy of Health Science (CAHS) Fellowship.

Induction into the CAHS as a Fellow is considered one of the highest honours within Canada’s academic community. CAHS Fellows, who serve as unpaid volunteers, are nominated by their institutions and peers and selected in a competitive process based on their internationally recognized leadership, academic performance, scientific creativity and willingness to serve.

Read more about Dr. Rosenberg and his incredible achievement in the USask article: [Celebrated USask medical researchers named Canadian Academy of Health Sciences fellows](#)

**Congratulations to Dr. Mark Inman** for being honoured with the Spirit of the College Award. **College of Medicine Team Achievement Award.** The Louis Horlick “Spirit of the College of Medicine Award” was established by the Alumni Association in honour of Dr. Louis Horlick, Professor Emeritus, Department of Medicine, for his contributions to the College of Medicine.

The annual award is presented to an individual associated with the college who has contributed over and above the call of duty and shows the spirit, dedication and enthusiasm that Dr. Horlick has exemplified over his career.

**New Podcast Alert!**

Department of Pediatrics’ Dr. Rupeena Purewal hosts **The Canadian Breakpoint.** A Canadian Infectious Diseases Podcast by Canadian Infectious Diseases Physicians. The latest episode is: CLEAR Registry-Canadian Leadership on antimicrobial rEal life usAge Registry Check out the episodes [here](#)!
Applying Quantum Computing in Human Health Research
A Revolutionary Saskatchewan-led Approach for Health Promotion and Disease Prevention
SYMPOSIUM AND WORKSHOP
November 9, 2023, 9:15 a.m. - 3:30 p.m.
Room 1B03 (First Floor B Wing), Health Sciences Building
University of Saskatchewan
A Mingling Minds Sponsored Event

in Association with the U of S Quantum Innovation and Health and Wellness Signature Areas

This symposium and workshop will bring together participants from a broad array of health sciences research disciplines, and representatives from health services delivery, healthcare administration, and health funding sectors. Renowned University of Saskatchewan quantum scientists will introduce basic information about the revolutionary power quantum computing can have for analyzing data to guide innovative health promotion and disease prevention strategies. This interactive event will provide opportunities to forge transdisciplinary collaborations and explore opportunities to access and apply quantum computing to human health information.

This in-person event is FREE but REGISTRATION IS REQUIRED before October 31, 2023.

Register Today!
For further information about this event please contact qc.health@usask.ca
## Coming Events

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<td><strong>QI power hour</strong></td>
<td>Advancing the Healthcare System Using Patient Reported Measures (QI Power Hour)</td>
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<td>Oct 24</td>
<td><strong>2023 Gairdner Lecture with Dr. Elizabeth Eisenhauer</strong></td>
<td>Seeking with Cure: Four Decades of Cancer Trials in Canada</td>
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<td>Oct 26</td>
<td><strong>Campus Conversations</strong></td>
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<td>Nov 2</td>
<td><strong>Pediatrics Grand Rounds</strong></td>
<td>– Dr. Hugh McMillan presents, “Gene replacement therapy: an emerging option in the treatment of neurological disease” (IN PERSON at Vivian Asher Lecture Theatre)</td>
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<td>Nov 9</td>
<td><strong>Applying Quantum Computing in Human Health Research</strong></td>
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New USask research chair focuses on improving Indigenous children’s wellness

Newly appointed Tier 2 Canada Research Chair (CRC) Dr. Wendie Marks (PhD) at the University of Saskatchewan (USask) is investigating links between obesity in Indigenous children and their exposure to environmental factors during pre-conception, prenatal, or early postpartum periods.

Read the full story Here!

Save the Date: Child Health Research Trainee Day

Department of Pediatrics
Child Health Research Trainee Day
Please join us on Thursday April 18th, 2024 for this research symposium featuring presentations from residents, graduate students, post-doctoral fellows, and undergraduate students engaged in child health research at USask.

Abstracts Due Friday March 29th, 2024
For questions and/or submissions please email them to tova.dybvig@usask.ca

The Children’s Health Research Trust Fund (CHRTF) was established in 1983 to help raise funds to support child health research at the University of Saskatchewan. As all donated funds are endowed, the CHRTF has continued to grow to become an important part in helping advance research in the Department of Pediatrics. For further information about the CHRTF and to donate: https://donate.usask.ca/online/chrtf.php

Our Partners:
The Jim Pattison Children’s Hospital has historically provided strong support for child health research in Saskatchewan. The recent $50 million donation from Jim Pattison allows for a steady stream of revenue to help meet research and programming needs for generations to come. Groundbreaking opportunities for pediatric researchers in Saskatchewan are on the horizon!

Contact us
For more information about The Department of Pediatrics Research, SPRING, or to contribute content to the Department of Pediatrics Research Report, please contact: Tova Dybvig Department of Pediatrics Email:Tova.dybvig@usask.ca