2020/21 ENDOWMENT REPORT

in support of the **Centre for Molecular Medicine and Therapeutics at BC Children's Hospital Research Institute**

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THANK YOU FOR CREATING A LEGACY

Thank you for your generosity, thoughtfulness and vision in establishing the Ed Turner Memorial Endowment for Medical Genetics. Endowments provide guaranteed support that enables the pioneering work of researchers, helps to train the next generation of health care professionals, and ensures the best possible care for children today and in the future.

Genetics underlies most childhood diseases, from rare single-gene disorders to more common diseases—like diabetes—that involve the complex interaction of many genes. The Centre for Molecular Medicine & Therapeutics (CMMT) is home to 11 principal investigators focused on understanding the roles of genetics and the environment on human health and disease. Researchers there pursue a better understanding of the genetic basis of human disease, from genes and molecules to cells and tissues. A key objective of their research and discovery efforts is to develop new genetic and cellular therapies for the treatment of childhood diseases.

The CMMT's location on the BC Children's Hospital campus means investigators can work closely with clinician and scientist colleagues at the Hospital and Research Institute to advance discoveries of biomedical tools and therapies for children impacted by genetic diseases. This closely connected community is committed to ensuring that children and families in BC and around the world can enjoy bright and healthy futures. Thank you for supporting this important research.

PROGRAM UPDATE

In July 2019, Dr. Bruce Verchere officially took the reins as Director of the Centre for Molecular Medicine & Therapeutics. Dr. Verchere, who you'll meet later in this report, brings his passion for child health research to the role after more than two decades leading, championing and promoting diabetes research at the Canucks for Kids Fund (CFKF) Childhood Diabetes Laboratories at BC Children's Hospital. He has hit the ground running, working closely with the CMMT's researchers, trainees, and staff to develop new initiatives and build community.

HELPING FUTURE RESEARCH STARS

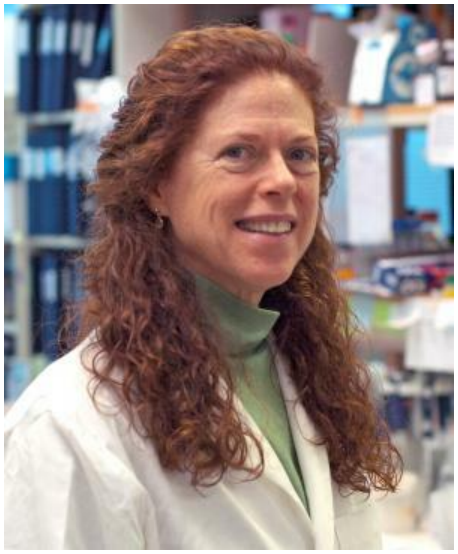
One of the CMMT's objectives is to empower its graduate students and post-doctoral fellows to work together to enhance the training experience and sense of community for trainees. Today, a flourishing CMMT Trainee Committee is comprised of nine early career researchers from eight different labs. This group has initiated and organized several exciting events, such as:

- In October 2020, three trainees presented at the virtual **Gairdner Foundation Symposium**, engaging high school students from around the province.
- Trainees played a key role in the first **CMMT Spring Symposium** in March 2021. This virtual event saw a noteworthy 170 registered attendees from 92 schools across the province.
- The CMMT Trainee Committee also created and coordinates a new seminar series, **CMMTalks**, which hosts two presentations monthly throughout the year.

The CMMT has a proud, 25-year history of research and discovery that is changing the lives of children and families. Thank you for your support in helping to build on this remarkable history and continue the Centre's vital work to further understanding of the genetic, epigenetic and environmental causes of disease.

RESEARCH SPOTLIGHT: SEEING THE FUTURE

Ground breaking research into the development of treatment for a rare form of congenital blindness is currently underway in Dr. Elizabeth Simpson's lab at the CMMT. It is also inspiring budding scientists to pursue a career in research.



Aniridia, a rare genetic eye disorder caused by mutations, results in the complete or partial absence of the iris (the coloured part of the eye) and varied levels of vision loss or impairment. Most children with aniridia have low vision starting at birth and may develop blindness and other eye-related problems, such as cataracts and glaucoma, in their teens or adulthood.

"There are currently no long-term vision-saving therapies or cures for aniridia, meaning that new treatment strategies are needed," said principal investigator Dr. Elizabeth M. Simpson.

Dr. Simpson's team first developed a model that can detect the variant causing the disease, then developed a CRISPR gene editing strategy that was used to correct the mutation in the gene sequence—thereby restoring vision. The model will be useful further research of this novel treatment and, importantly, the development of other therapeutic approaches.

These findings represent crucial steps toward a future cure for aniridia in children.

What is CRISPR?

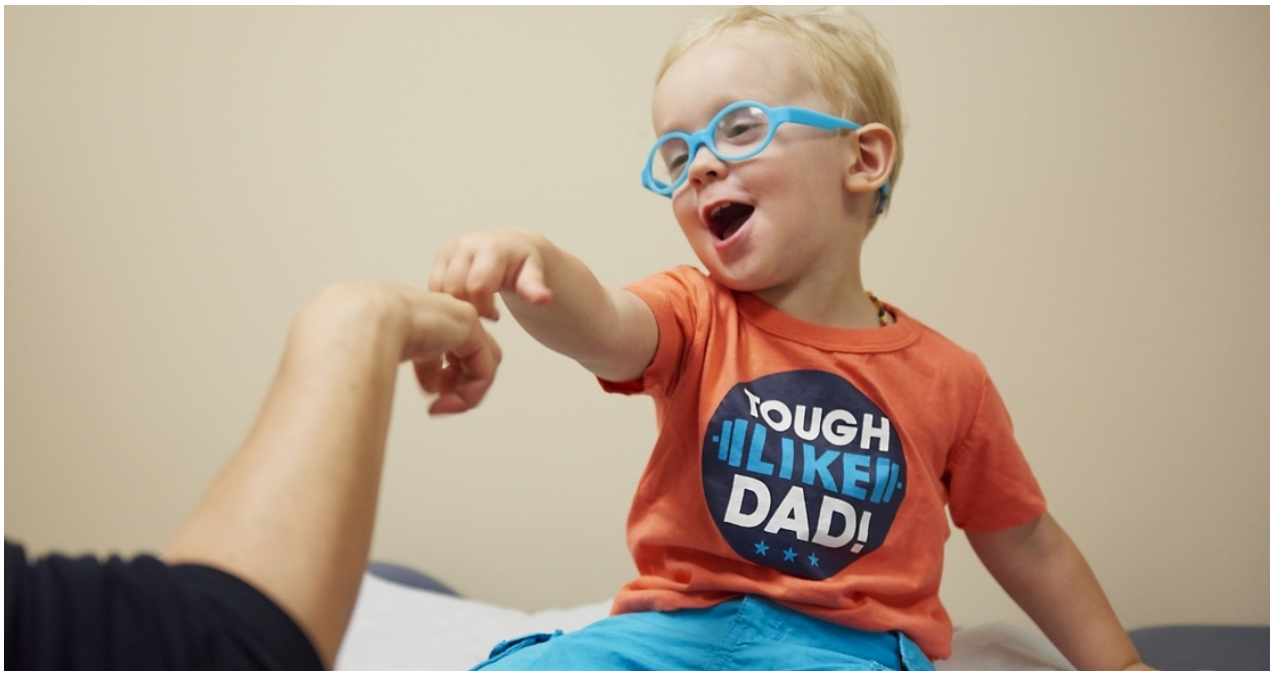
Gene therapy is an exciting new class of precision medicine where DNA is delivered into a patient's cells to treat disease. CRISPR is a powerful new gene therapy that is like surgery for the gene—directly editing and correcting gene mutations. Through CRISPR, researchers can create specific treatments for children with genetic diseases.

"My team and I were thrilled to explore this new approach to the treatment of childhood blindness. Our success will help strengthen the foundation for gene-editing approaches in treating children with aniridia and other genetic diseases," said Dr. Simpson.

Recently, Dr. Simpson participated in a podcast discussing her research and the role of CRISPR and genome editing therapies focused on the search for a cure. Notably, outreach like this is central to the CMMT as it was set up and to its mission. It is one of the three components upon which staff are evaluated, namely publications, funding, and outreach. "Obviously publications and funding are critical. But outreach is their equal," Dr. Simpson said. "It may not be easy or convenient but people make time for it. Doing the podcast was fun." There are many formats and delivery methods for getting people excited about research, from newsletters and blogs, tutoring, organizing or judging science fairs, even volunteering at Science World.

Inspiring the next generation of scientists as well as those who have recently embarked or are well-established on their scientific paths, is critical. After all, everything begins with inspiration and it's through outreach that inspiration can take root, treatments be found and, by way of example, vaccines developed.

Outreach is part of being a scientist at all levels. "It's good for your communication skills and you learn, in some cases, how to speak well to the lay audience," Dr. Simpson said. "You're actually practicing your scientific skills." And, in so doing, lighting a spark.



LEADERSHIP SPOTLIGHT: CMMT DIRECTOR DR. BRUCE VERCHERE

"Research at the CMMT has the potential to uncover the underlying causes of genetic diseases, leading to new therapies that can transform the lives of kids and families. Your support of the CMMT and BC Children's makes this life-changing research possible. You are helping to create a truly remarkable research environment where clinicians and scientists work together with patients to decipher the complex causes of genetic diseases in kids and families. Thank you."

– Dr. Bruce Verchere, Director, Centre for Molecular Medicine and Therapeutics

WHAT ATTRACTED YOU TO THE ROLE AS DIRECTOR OF THE CMMT?

The CMMT has been a world leader for research in genetic medicine and its translation for the past 25 years. This position is an exciting opportunity to help the Centre grow and further its research excellence and impact on the health of kids. Our research, aimed at understanding the genetic basis of rare disease and translating those findings into new prevention, diagnosis and treatment strategies, has huge potential to benefit kids. Personally, this opportunity came at a good time for me. After leading the CFKF Childhood Diabetes Laboratories for more than 15 years, I was ready for a new challenge.

WHAT HAVE YOU ACCOMPLISHED SO FAR?

One of the first things I did was meet with as many people as possible, including researchers, trainees, and staff, to learn all I could about the CMMT. I wanted to get a sense of what the Centre means to its members, where they would like to see the CMMT in a few years, and to identify where there were gaps and issues that needed attention. I spent a lot of our downtime during the pandemic in 2020 continuing to learn about the Centre's history, research, and people. We began a strategic planning process early in 2021 which is nearing completion, and have identified several important strategic initiatives. One key initiative is growth through internal and external recruitment. This represents a big challenge but is also a fantastic opportunity to build on the CMMT's remarkable history of research success and help take it to even greater heights.

WHAT ARE YOU ENJOYING ABOUT YOUR ROLE?

It has been incredible learning about all the fantastic research going on in the CMMT, mostly through meetings with trainees and faculty and attending seminars and 'chalk talks.' Before taking on this role, I was aware of the great research happening there, but didn't fully appreciate what a diverse and dedicated group of outstanding scientists there are. My meetings with the Centre's trainees, especially, have made me appreciate that the future is very bright for child health and genetics research at the CMMT and BC Children's. I'm excited to have this opportunity to make an impact and support this research by removing barriers and helping to enable growth and further collaboration. The potential for life-changing research into the genetic causes of childhood disease are immense and I am excited about what the next few years will bring.

HOW HAS YOUR RESEARCH CHANGED SINCE TAKING ON THIS ROLE?

In just two years I have started new collaborations with most of the other CMMT labs, taking our research in directions I had not previously considered. I have learned much about how genetic and computational biology approaches can impact my research, and am starting to use these approaches in collaboration with other researchers, already with some exciting results. I am still very much a diabetes researcher—I have been my entire career—trying to understand why insulin-producing beta cells stop working in kids with diabetes, and my interactions with the other labs have opened doors to exciting new approaches to my research.

THANK YOU

Your support is helping enable the genetic research that will shape the future of children's health. Thanks to your generosity, the CMMT continues to excel under new leadership, with its scientists at the forefront of this leading-edge research, fostering collaborations and creating an environment where the next generation of genetic researchers can grow and thrive. Your endowment has created a remarkable legacy that will be felt for generations. Thank you.