POGO-FUNDED RESEARCH

Identifying Children at Increased Risk for a Cancer Predisposition Syndrome

Approximately 10% of children diagnosed with cancer have an underlying cancer predisposition syndrome (syndrome), a condition (often inherited) that increases their risk of developing one or many cancers throughout their life. Genetic testing can be used to identify children with a syndrome, but to avoid unnecessary testing and possible delays, physicians need a way to determine which children to test.

There are over 125 syndromes known to be associated with cancer in children. In some cases, knowing whether a child has a specific syndrome will change the treatment. Identifying a syndrome also allows the healthcare team to develop a care plan to monitor the child for future cancers once treatment is complete, and to recommend monitoring for siblings.

In 2017, Dr. Catherine Goudie was awarded a POGO fellowship for her project "Identifying Children at Increased Risk for a Cancer Predisposition Syndrome: The McGill Interactive Pediatric Oncogenetic Guidelines." Overseen by principal investigators Dr. David Malkin and Dr. William Foulkes, the project aims to develop an e-Health tool that doctors can use to help them decide which children should be referred for genetic testing.

Prior to becoming a POGO Fellow, Dr. Goudie and her team in Montreal spent three years building approximately 90 algorithms for each type of childhood cancer. These yes/no questions relate to items such as the age of the child, features of the tumour and family history. The first version of the tumour algorithms was reviewed by genetic and oncology experts from Canada, the United States and the United Kingdom.

The aim of the POGO-funded research was to support Dr. Goudie in her work to evaluate the performance of the tool with children at 11 Canadian hospitals. The evaluation was split into two streams.

"Our first priority was making sure this tool would do no harm," says Dr. Goudie. "In order to do this, we looked at patients in Canada who have already been diagnosed with a cancer predisposition syndrome and who developed a cancer in childhood, meaning their physicians successfully identified the syndrome without our tool. We confirmed that our algorithms would have identified these children for genetic referrals at the time of their cancer diagnosis, therefore confirming that the tool performs at least as well as clinicians have."

The second stream of the evaluation is ongoing and leverages precision medicine research at The Hospital for Sick Children and other hospitals in Canada and the US. These institutions are doing comprehensive genetic sequencing on pediatric cancer patients, meaning that all children undergo genetic evaluation regardless of whether they are suspected to have a syndrome.

"We will test our algorithm on the medical profiles of these children to determine if the tool indicates that the child should be referred for genetic assessment. We will then compare our results with those obtained from the patients who participated in the comprehensive genetic sequencing. Therefore, we are prospectively testing the performance of our tool."

Dr. Goudie explains that, in most hospitals worldwide, the facilities, human resources, infrastructure, expertise, or funding to offer comprehensive genetic testing in all kids diagnosed with cancer are not available. Ultimately, the goal of the tool is to give children with a syndrome the opportunity to be diagnosed and treated appropriately. In addition, the tool can be used to educate medical professionals around the world.

"For someone young like me, who doesn't have a lot of experience, having POGO to support my work was a huge stepping stone. I am certain that POGO's support and the credibility that this organization has internationally, helped me get this next award in Quebec. I am really grateful for this opportunity." **– Dr. Catherine Goudie, MD, FRCPC**

Dr. Catherine Goudie received a prestigious clinical investigator award from the FRQ-S in Quebec and returned to Montreal in July 2018 to continue this research as a collaboration between the Montreal Children's Hospital and The Hospital for Sick Children. The FRQ-S is a government organization that, annually, funds select clinician-investigators in Quebec to do health research.

STATS + FACTS

In addition to the Research Fellowship Program, POGO offers annual Seed Funding Grants of up to $15,000 to facilitate or support feasibility studies, the development of preliminary data to enable applications to other granting agencies or self-contained studies. Studies funded this year were:

Eric Bouffet, MD, FRCPC
Parent-Child Communication When a Child Has a Life-Threatening Illness

Denise Mills, MN, NP Pediatrics
Sue Zupanec, MN, NP Pediatrics
Improving Quality and Consistency in Family Education Prior to First Discharge Following a Pediatric Cancer Diagnosis

Lindsay Jibb, RN, PhD
Needs Assessment Focused on Home-Based Care for Children with Cancer: A Qualitative Descriptive Study Using the Perspectives of Children, Parents and Healthcare Providers

Underpinning the work of the POGO Research Unit is the POGO childhood cancer database. Unmatched by any other in the world, POGONIS◆ contains more than 30 years of standardized and comprehensive information on diagnosis, treatment, complications and long-term outcomes of children diagnosed with cancer in Ontario.

IN FISCAL 2018, DATA FROM POGONIS SUPPORTED

37 **INVESTIGATORS** across Ontario

\+ and in addition supported

13 **PROVINCIAL/ NATIONAL** research projects with external investigators

THANK YOU

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