a place of mind

STEWARDSHIP REPORT

PRIEST FAMILY FUND FOR MORQUIO B

2015-2016 IMPACT REPORT

Prepared by the UBC Faculty of Medicine

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FACULTY OF MEDICINE

Overview

Your support makes a vast difference to the success of our research and the health of our community.

Thank you.

COMMITMENT TO FINDING A CURE

In 2009, Dan and Tina Priest discovered that their son, Stephen, has a rare condition affecting the development of his bones and joints. As Stephen grows, his body is becoming less mobile and increasingly fragile, so that a small slip or bump may be life-threatening if nothing is done to surgically repair the joints between his bones. At the same time, this disease can lead to heart, lung, and hearing problems, and, in some cases, a decline in brain function.

The genetic illness is called mucopolysaccharidosis (MPS) IV B, or Morquio B, and is extremely rare. Currently, less than 100 people with this condition have been identified worldwide, but many more patients may be living with painful bone deformities without knowing their correct diagnosis. Since it is so rare, research into possible therapies and cures to Morquio B is equally rare, and people living with its devastating consequences currently have no access to treatment other than surgery. Even then, surgical procedures can be extremely difficult, since the bones in people with Morquio B are not only abnormally shaped but also heal and grow unlike regular bones. Most patients need multiple bone surgeries during their lives.

To help Stephen and people like him around the world, the Priests have dedicated themselves to creating a solution by raising funds and awareness for improved treatments and, one day, a cure. Over the past six years, the Priests have raised close to $825,000 to support critical Morquio B research led by Dr. Sylvia Stockler, Stephen's physician and Professor and Head of the Division of Biochemical Diseases in the UBC Department of Pediatrics. Dr. Stockler and her team at British Columbia Women's and Children's Hospital are working with a growing international group of experts to expand knowledge and care options for people with Morquio B. Dr. Eduard Paschke from the Medical University of Graz, Austria, who was involved in the discovery of the disease's genetic background in the 1970s, and Dr. Clara van Karnebeek, a rare disease researcher at the Child and Family Research Institute of BC Children's Hospital, are working alongside Dr. Stockler. Both investigators were among the initial founders of a research program dedicated to finding people with Morquio B across the globe. The project aims to collect medical data from these patients to lay the foundation for new treatments.

As these researchers dedicate their expertise to understanding Morquio B, their findings are leading to a brighter future for people like Stephen.

Accomplishments in 2015-2016

THE MORQUIO B PATIENT REGISTRY

In October 2012, Dr. Stockler and her team created an online patient registry at morquiob.org to gather information from people worldwide with Morquio B. The registry is the first international compilation of demographic and disease-specific data for Morquio B, and will lay the foundation for new approaches and discoveries in research. Over the past year, the MorquioBetter team's efforts to promote the registry have been highly successful, and the number of registrants has grown to 30 people worldwide, representing one third of all people with Morquio B. In addition, many more patients have expressed interest in participating in the registry. The rich database is becoming instrumental to expanding our knowledge about the causes, symptoms, and potential treatments for Morquio B.

A REMARKABLE GIFT

We are pleased to share an inspirational story about a recent generous estate gift, contributed by Elizabeth Done to the Priest Family Fund for Morquio B. This support, in combination with your generosity, is increasing the capacity of Morquio B research for Dr. Stockler and her team.

Giving to Save Lives

Elizabeth (Betty) Done (April 12, 1940 to April 27, 2015)

Betty Done was an energetic and generous lady with an exceptional love for her two children and six grandchildren. She was known to tell heartwarming stories about her family, always with a twinkle in her eye and a broad smile. Two of Betty's grandchildren, Damien and Natasha, were diagnosed with Morquio A at a very young age. Betty enjoyed having these two grandchildren and their siblings over for visits, sleep overs, and baking sessions. She witnessed the struggles caused by Morquio A and became a committed donor to research for a treatment.

Sadly, on March 3, 2011, Damien lost his life to Morquio A at the young age of 18. Just a few years later, researchers developed enzyme replacement therapy to treat Morquio A. Natasha may be able to benefit from this advancement, and she recently applied for provincial funding to pursue the costly therapy.

Betty saw first-hand the exceptional progress researchers can make when supported by generous members of the community. She also understood the transformational impact of new treatments in saving and enhancing the lives of loved relatives and people of all ages. After getting to know the Priest family and watching their tireless efforts to raise funds for Morquio B, Betty decided to direct her support to Morquio B research. Before her passing, she also made the admirable decision to leave a gift in her will to advance this research at UBC. Betty donated with the hope that investigators will one day find a cure for Morquio B, enabling children like Stephen Priest to live longer, more fulfilled lives.

Accomplishments in 2015-2016

The Priest Family, Tina, Stephen, and Dan, with Dr. Stockler.

INVESTING IN NEW AND BETTER TREATMENTS

The MorquioBetter team is eager to identify and participate in the most promising international Morquio B research as they strive for improved treatments. The team continues to lead discussions with investigators and organizations across the globe to find current projects in the field that will both benefit from the robust database gathered through the Morquio B registry and add new advances and expertise to the project. In the midst of a dynamic environment of drug development for rare diseases, Dr. Stockler will assemble a think tank of leading physicians and researchers to identify the research projects with the greatest potential. Registry and scientific findings will continue to work hand-in-hand, as patient data inform possible drug targets, and drug testing occurs more effectively with ready collections of cells and patients.

The team is also seeking out rare disease foundations that support research in Morquio B and related diseases, as well as companies working on drugs to treat Morquio B, with the goal of pooling resources and expertise.

WORKING WITH CARE PROVIDERS

As research continues to move forward, a more immediate opportunity to improve the wellbeing of people with Morquio B may arise from partnering with the primary care providers who work with patients. Current treatment is limited to a series of surgeries, such as spinal fusion, carried out by orthopaedic surgeons to stabilize the bones and joints. The MorquioBetter team is developing strategies to teach care professionals like orthopaedic surgeons about advances in our understanding of Morquio B. By educating care specialists about disease characteristics, progression, and expectations, the current course of treatment can be enhanced to account for potential complications and improve care outcomes. Ultimately, our ability to share knowledge and findings among diverse groups of care takers will enhance the quality of life of people with Morquio B.

Thank You

Your support is helping people and families with Morquio B overcome the isolation and hopelessness of living with a rare disease by uniting these families with researchers across the globe to discover new therapies and a potential cure. Thanks to your generosity, experts in rare genetic diseases are able to dedicate their time and experience to advancing our knowledge about Morquio B. In the process, they are improving the independence and wellness of people like Stephen.

The MorquioBetter Team

While an international team of experts have been working together on this project, three scientists have taken a lead role in making the Morquio B registry a reality.

Dr. Sylvia Stockler, Principal Investigator

Dr. Stockler is Professor and Head of the Division of Biochemical Diseases in the UBC Department of Pediatrics and Program Director of Biomedical Diseases at BC Children's Hospital. She is a leading authority on rare diseases with a focus on treatable inborn errors of metabolism. She also serves as the project leader of TIDE BC, a collaborative care and research initiative whose goal is to provide timely diagnosis and treatments for children with treatable forms of intellectual disability.

Dr. Eduard Paschke, Co-Investigator

Dr. Paschke, Associate Professor, Department of Pediatrics at the Medical University of Graz, Austria, is an internationally renowned researcher who has dedicated most of his research to Morquio B disease and late onset GM-1-Gangliosidosis. His Laboratory of Metabolic Disorders has developed comprehensive biochemical and genetic methods to diagnose these and other disorders and functions as a reference laboratory for diagnosis of Morquio B disease in Europe and overseas.

Dr. Clara van Karnebeek, Co-Investigator

Dr. van Karnebeek holds a position as Clinical Assistant Professor in the UBC Department of Pediatrics. Her work is focused on diagnosing and finding innovative treatments for patients with intellectual disabilities. She is the recipient of the Laura McRae Award for Excellence in Pediatrics from the BC College of Physicians and Surgeons, the 2012 Digital Health Innovation Summit Award, and the prestigious Maud Menten Award presented by the Canadian Institutes of Health Research.