a place of mind

STEWARDSHIP REPORT

PRIEST FAMILY FUND FOR MORQUIO B

2014-2015 IMPACT REPORT

Prepared by the UBC Faculty of Medicine

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 currently known to affect fewer than 100 people worldwide. 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.

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 five years, the Priests have raised close to $280,000 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 is working alongside Dr. Clara van Karnebeek, a Clinical Assistant Professor experienced in rare disease research at UBC Pediatrics, and Dr. Eduard Paschke from the Medical University of Graz, Austria—one of the few scientists worldwide engaged in Morquio B research.

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

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Accomplishments in 2014-2015

Capilano University Musical Theatre students performing at the Black and White Masquerade

A BLACK AND WHITE MASQUERADE

On October 18, 2014, the Priest family hosted an exciting and lively Black and White Masquerade to celebrate the accomplishments of Morquio B research to date and to raise additional funds for ongoing investigation. The evening was a delightful success and helped raise more than $40,000 to support Morquio B research.

THE MORQUIOBETTER PROJECT: CREATING AN INTERNATIONAL PATIENT REGISTRY

To better understand the symptoms and underlying causes of Morquio B, Dr. Stockler and her team set up an online patient registry on morquiob.org in October 2012. The registry consists of two databases, one that will collect basic patient information such as demographics and physician contact information, and a second that will compile more extensive information such as age of diagnosis, clinical symptoms, and genetic information. Dr. Stockler and her team have been widely promoting this registry through their websites, other rare disease platforms, and physicians around the world. Recently, the commitment of the Priests has enabled Dr. Stockler to expand and refine the questions on the registry to determine the range of ailments and most likely genetic factors involved in the illness. With this information, we will be closer to understanding the genetic changes that lead to Morquio B and how to help prevent or reverse these mutations.

Thanks to the MorquioBetter Project, people with Morquio B from around the world have begun registering their physical, psychological, and cognitive conditions, and so far, 30 people from countries including Brazil, Germany, Austria, Poland, and the USA have joined the registry. The morquiob.org site is quickly gaining ground, and international rates of enrollment are accelerating. In time, the registry aims to capture all cases of Morquio B worldwide. While providing scientists with a wealth of knowledge, this information could also create a critical mass of data that may help to raise government and industry awareness of Morquio B. The registry will also facilitate clinical trials by encouraging communication between researchers, patients, and physicians.

The registry aims to capture all cases of Morquio B worldwide. The patient histories can help examine which current treatments may be most effective and facilitate future clinical trials.

With 30 registrants, Dr. Stockler's team is ready to begin analyzing the data collected. The information they gain by organizing and categorizing registry responses has great potential to lead new clinical trials for treatment options. Comparing the genetic differences that cause Morquio B across a large number of patients will help to improve our understanding of each patient's

long-term prognosis. Although in Stephen's case physicians can already predict that he will not have severe brain involvement, for many other children with particular gene mutations, the threat of additional brain complications can only be answered during the long course of the disease. In addition to this type of data, the registry will help provide benchmarks to track the progress and effectiveness of treatment options as they are trialled.

INTERNATIONAL COLLABORATION FOR AN INTERNATIONAL CURE

The Priest Family: Tina, Stephen, and Dan

Currently, Dr. Stockler and her team are establishing an exciting new collaboration with a leading-edge research group in Europe that is also dedicated to finding a cure for Morquio B. This group, led by Dr. Tanja Wrodnigg, Professor of Organic Chemistry at the Graz University of Technology, and Dr. Eduard Paschke at the Graz Medical University, has successfully developed chemical substances with the potential to correct the genetic defect leading to Morquio B. This group has a collection of cells from patients around the world, which allows them to test the safety and effectiveness of their substances and eventually develop these substances into promising new treatments. Based on encouraging results from initial experiments, the group is now working to refine molecules for the most beneficial therapeutic effect. Funding from the Priest Family Fund for Morquio B will support this ground-breaking research.

Dr. Paschke is also working to apply data from the Morquio B patient registry to discoveries from his experiments. Uniting the large database of patient information from the registry with drug testing results will enable Morquio B researchers to proactively test new therapies in patients. The 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 pools of cells and patients.

This collaboration is sure to deepen our understanding of the disease and facilitate faster, more reliable trials leading to a future cure.

Thank You

Your support is uniting families around the world in their search for a cure. Each contribution brings us closer to uncovering the causes of Morquio B and discovering a solution that will help end the suffering brought about by this rare disease.

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 Institute of Health Research.