a place of mind

STEWARDSHIP REPORT

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

Thank you.

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PRIEST FAMILY FUND FOR MORQUIO B

2012-2013 IMPACT REPORT
Prepared by the UBC Faculty of Medicine

COMMITMENT TO FINDING A CURE

In 2009, Dan and Tina Priest received the news that their son, Stephen, had a genetic condition called mucopolysaccharidosis (MPS) IV B, or Morquio B. This illness has no known cure and is extremely rare. It leads to problems in bone and joint formation and function, causing reduced physical mobility and pain and often requiring multiple orthopaedic surgeries. Among other symptoms, it can lead to heart, lung and hearing problems.

Since the disease is so rare, there has been little investment by governments and pharmaceutical companies to search for a treatment. Instead of giving up hope, the Priests decided to focus on finding a solution by raising money and awareness to help Stephen and those like him.

The Priests have raised more than $210,000 for Morquio B research since 2010. Dr. Sylvia Stockler, Stephen's physician and Professor and Head of the Division of Biochemical Diseases in the UBC Department of Pediatrics, leads the research, working in collaboration with 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.

With research underway, the important work towards a better understanding and improved outcomes has begun.

THANK YOU

The Faculty of Medicine sincerely appreciates your generous support of Morquio B research. Your contributions have fuelled an important advancement in the treatment of this serious illness. Thank you for your dedication to improving the health of individuals around the world diagnosed with this rare disease.

Accomplishments in 2012-2013

The Priest Family: Tina, Stephen and Dan

THE MorquioBetter PROJECT: CREATING AN INTERNATIONAL REGISTRY

Dr. Stockler and her team determined that the first step toward improving outcomes for Morquio B patients and families would be to locate as many patients as possible and gather data about their disease histories. Knowing about patients' physical, psychological and cognitive functions at the different stages and ages of their lives is important to understand the spectrum of severity of this condition. Understanding this natural history is extremely important for rare diseases like Morquio B as this is the first and crucial step for developing new treatments.

With funding raised by the Priest Family, the MorqioBetter Project — a comprehensive international patient registry — was launched in October 2012. Its implementation now well underway, Dr. Stockler and her colleagues have made tremendous progress in raising awareness of Morquio B and collecting the international data that will provide the foundation for research that could lead to new treatments. As a direct result of the Priest family's efforts and the substantial support of donors, this initiative has established a global community of families living with Morquio B and inspired new hope that this illness might one day be cured.

GATHERING IMPORTANT DATA

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.

The Morquio B registry is designed to provide scientists with a better understanding of how both Morquio B and a related condition called late-onset GM-1-Gangliosidosis progress over time. As a genetic condition, Morquio B is caused by a mutation on the GLB1 gene. Late-onset GM-1-Gangliosidosis is caused by a different mutation on the same gene. However, in addition to bone and organ disease, late-onset GM-1-Gangliosidosis can have devastating effects on the brain resulting in cognitive decline and severe handicap. Compiling information on both illnesses will provide researchers with valuable insights into how these genetic variations affect individuals with these diseases over time.

In addition to collecting patient data of the particular progression of these diseases in each individual, the registry will gather information on treatments they have undergone, providing benchmarks to help researchers examine which treatments may be most effective.

The registry aims to capture all the cases of Morquio B worldwide. While it will provide 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 future clinical trials by enabling communication between researchers, patients and physicians.

Progress and Vision

Stephen is the first patient enrolled in the new registry. International patients will be added in 2014.

Stephen Priest and friend

RECENT PROGRESS

To overcome the obstacle of geographic isolation of single patients, Dr. Stockler and her co-investigators envisioned the use of a secure, online patient medical questionnaire that could be accessed anywhere. This questionnaire would enable the MorquioBetter study team to compile valuable patient information, while finding sufficient numbers of patients to conduct clinical trials once treatments become available. Dr. Stockler convened a consortium of Morquio B experts from a variety of countries around the world, including Austria, Brazil, Canada, Germany, Greece, Italy, Japan, Spain, and the USA to obtain consensus about the formulation of the questionnaire that will be used to gather relevant information on patients with Morquio B.

In February 2013, Dr. Stockler and her colleagues presented information on the registry at the World Lysosomal Disease Congress in Orlando, Florida. In September 2013, the same group presented the registry at the Annual Meeting of the European Study Group for Lysosomal Diseases in Graz, Austria, to engage most influential stakeholders in Europe. Both conferences provided an opportunity to obtain input from stakeholders and to create a most comprehensive version of the questionnaire capturing a variety of clinical, biochemical and genetic patient data.

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. Vancouver has been designated the central coordinating study site.

Stephen was the first patient enrolled in the registry. Enrolment of patients from international participating sites will start in 2014.

NEXT STEPS

Dr. Stockler and her team will promote the registry widely on the morquiob.org website, other rare disease platforms and through direct communication with physicians and scientists working in the MPS community. Patients and physicians will be invited to complete the questionnaire, and their responses will be added to the registry.

Once sufficient information has been compiled, Dr. Stockler and her colleagues will be able to identify key research questions that will bring them closer to new treatments, and possibly a cure, for Morquio B. The research team also plans to use information in the registry to identify the major health issues affecting patients with Morquio B — which are still not fully understood — and draft clinical guidelines on how to manage the condition based on existing knowledge. These guidelines will ensure physicians around the world have the support and information they need to provide effective patient care and will help to improve the quality of life of those suffering from this condition.

The MorquioBetter Team

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.

Drs. Sylvia Stockler, Eduard Paschke and Clara van Karnebeek

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.