40 Year Medical Mystery Solved

Hereditary Mutated Red Blood Cell Found in Family Tree, Thanks to Manitoba Medical Legacy

Winnipeg, Manitoba September 10, 2012 – A decades old mystery in genetics has been solved by a team of Manitoba researchers from CancerCare Manitoba (CCMB), and the University of Manitoba (U of M). With support from the Manitoba Medical Services Foundation, the project, led by CCMB's Dr. Ryan Zarychanski, has identified a genetic mutation responsible for hereditary xerocytosis, a disorder that causes the rapid destruction of red blood cells, one that has baffled researchers around the world.

The mystery began 40 years ago, when the first Manitoba patient was diagnosed with a blood disorder that resulted in a significantly reduced lifespan of red blood cells. The cell breakdown left patients prone to jaundice, gallstones, enlargement of the spleen, low hemoglobin, and iron overload, which can be harmful to the liver, heart and other organs.

With further investigation by Dr. Lyonel Israels, CancerCare Manitoba's founding father, it became apparent the condition was hereditary, as over half of the first patient's family was affected. Through the years, additional family members received treatment from CCMB's hematology experts, which includes Dr. Israels' own daughter, Dr. Sarah Israels.

In 2010, CCMB's Dr. Ryan Zarychanski, also a U of M assistant professor, along with Dr. Don Houston (who trained Dr. Zarychanski as a hematologist) and a team from CCMB and the University of Manitoba, initiated a study to get to the bottom of the disorder. Given the hereditary nature of the disease, the researchers sought out as much of the affected family's input as possible, which led to some unique fact finding.

"The project started with a family reunion in western Manitoba. Approximately 130 family members of the original patient provided blood samples and medical histories at a lab we set up in a rural community centre," says Dr. Ryan Zarychanski, the study's principal investigator. "From there, in collaboration with the University of Manitoba, and with support from Yale University in the United

States, we used a series of sophisticated DNA techniques and were able to pinpoint the exact genetic mutation responsible for the disorder."

The team's findings garnered the cover story of Blood, the world's top medical journal on blood disorders. The mutated protein found in the Manitoba family by Manitoba researchers is likely to help doctors to understand other inherited red blood cell disorders that affect families around the world.

"This study highlights the importance of continued investment in research to improve care in Manitoba," said Dr. Dhali Dhaliwal, president and CEO of CancerCare Manitoba. "With support from the Manitoba Medical Services Foundation, these Manitoba researchers worked together to solve a medical mystery affecting Manitobans, which will eventually translate into improved care for patients with blood disorders far beyond this province."

"It is very rewarding to see the collaborations and the knowledge that have been generated from this investment in research. The ongoing support of the MMSF by the Manitoba Blue Cross enables this funding for education and research targeted to new investigators in Manitoba", said Dr. Greg Hammond, Executive Director of the Manitoba Medical Service Foundation.

Along with the benefits of two generations of expertise in hematology, the study is helping train the next generation of Manitoba doctors. The study team included University of Manitoba medical student Brett Houston, who has won provincial, national and international awards related to this gene discovery.

"I commend Dr. Zarychanski and his colleagues on this groundbreaking discovery," said Dr. Brian Postl, Dean of Medicine, University of Manitoba. "This project demonstrates an outstanding example of collaborative research, and highlights our faculty's commitment to training clinician scientists. The university and medical faculty will continue to invest in and encourage research collaborations and student research opportunities."

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