CENTRE FOR GENOMICS ENHANCED MEDICINE

Patient Impact Story

The Gift of Mowing the Lawn

Growing up most of us dream of owning a home. But for people like Ryan, who was born with Fabry disease, the burden of upkeep often makes home ownership unattainable.

Fabry disease is caused by a change in the GLA gene which decreases production of an enzyme to break down certain fatty materials in the body. "Fabry disease is inherited," says Dr. Michael West, a renowned nephrologist and researcher with the Center for Genomics Enhanced Medicine (CGEM). "It starts in childhood with severe pain in the hands and feet due to nerve damage; from there the disease causes decreased sweating and leads to progressive kidney disease, heart disease, and strokes all before age 55."

For Ryan the burning pain in his hands and feet started when he was six. He recalls visiting doctors who said there was nothing they could do. But that answer was unacceptable to Dr. West, particularly as Nova Scotia had such a high prevalence of Fabry Disease because of a founder effect. "Geneticists have traced Fabry disease back to a common ancestor who arrived here in 1750 and it is her descendants that have this condition," continues Dr. West.

Determined to do better for his patients, Dr. West became chief investigator for a multicentre, phase 3 clinical trial studying enzyme replacement therapy (ERT) in Fabry disease. The timing was ideal because Ryan, now in his mid-twenties, had started showing signs of organ damage, with decreased kidney function and a thickened heart.

With the encouragement of his then girlfriend, now wife, Ryan agreed to ERT with intravenous drug infusions every two weeks. Within six months, he went from having pain every day to feeling normal. Ryan's medical check-ups revealed the enzyme was working, breaking down the fatty deposits and stabilizing the organ damage.

> *"ERT has been life altering for me," Ryan says. "Before I would stay inside all summer long, unable to cope with the heat. Today, my wife and I have bought our first house and I am able to mow my lawn."*

"The success of the clinical trial means Fabry patients are living longer and ERT is now covered under provincial drug programs across Canada," says Dr. West. He cautions that this therapy, while effective at managing Fabry disease, is not a cure. It requires patients to receive treatment every two weeks and costs around $300,000 per patient per year.

Scientific advances made by a national consortium, in collaboration with CGEM researchers, have led to the launch of a new phase 1 clinical trial testing a gene transfer therapy. With funding from the Canadian Institutes of Health Research, scientists have re-engineered a virus to deliver a normal functioning GLA gene into the cells of Fabry patients. This could potentially be a more permanent and effective treatment for Fabry disease. Dr. West is leading Halifax segment of this national trial and currently recruiting patients. "This trial is a first in Canada, a first in the world, and that's pretty exciting," he says.

Ryan shares Dr. West's excitement in the promise that gene therapy holds saying, "Just knowing there are other treatments coming down the pipe, means I can plan for the future."

Photo Credit: Nick Dollimount

In partnership with: DALHOUSIE UNIVERSITY, IWK Health Centre, nova scotia health authority

CGEM Contact: DR. MICHAEL WEST
EMAIL: info@cgem.ca TEL: 902-473-4023