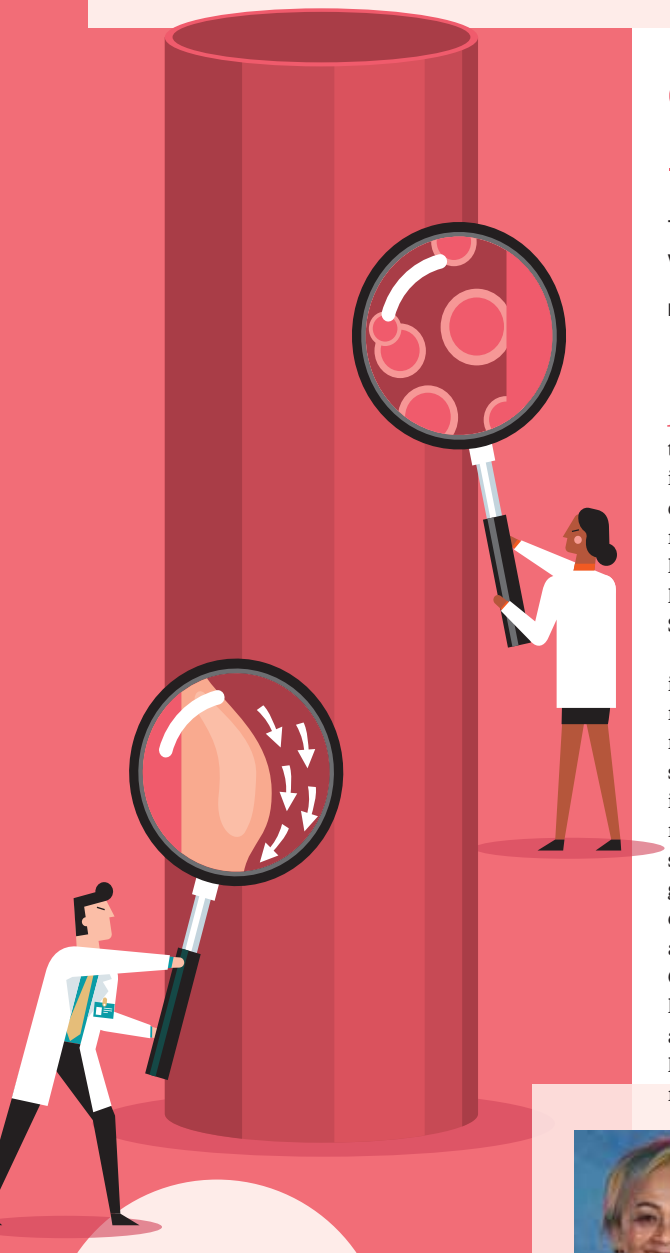
Rare Diseases



Conquering Rare Disease — Even In the Era of COVID

The long-awaited launch of Canada's Rare Disease Drug Strategy will help get therapies to people with rare diseases faster.

Dr. Durhane Wong-Rieger

As Canada prepares to pivot from reacting to COVID-19 as an unpredictable pandemic crisis to living with COVID-19 as a manageable influenza-like endemic, the federal government is preparing to make an equally momentous pivot in treating Canadians living with rare diseases, with the launch of Canada's Rare Disease Drug Strategy in 2022.

The most important aspect of this initiative isn't the government's commitment of $1 billion to set up the program, nor is it the $500 million annual budget to sustain it. What constitutes a seismic shift is Canada's recognition that therapies to meet the unmet needs of small and very small patient populations with severe, progressive, life-threatening, and untreated disorders demand unique customized access pathways. For over 40 years, Canada (along with most other countries) has applied to rare drugs the same criteria and tools that were designed for assessing large population drugs, many of which are nth generation incremental improvements on existing therapies with proven impacts on well-characterized conditions.

Dr. Durhane Wong-Rieger
President & CEO, Canadian Organization for Rare Disorders

The slow pathway to gaining access

It's no wonder that rare disease drugs were tried and found wanting. The clinical trials were too small or too short — they lacked placebo or proper control groups, relied on biomarkers rather than confirmatory outcome measures, and yielded an incremental cost-effectiveness ratio that was highly uncertain and far above the standard cut-off. In fact, it's surprising that any rare disease drug at all made it through this highly inappropriate gamut.

Over the years, many countries have developed assessment tools, criteria, and separate access pathways that are more flexible or specific to rare therapies. Canada chose not to follow suit. Not surprisingly, rare disease patients in Canada have lagged than those in many other developed and developing countries in gaining access to approved innovative therapies.

Progress for the rare disease community

For nearly two years, the Canadian Organization for Rare Disorders (CORD), the alliance of rare disease patient organizations, has led the multi-stakeholder consultations on the national Rare Disease Drug Strategy and gained consensus on key aspects, including vision, governance, infrastructure, operating principles, core access pathways, success factors, and the role of patients.

For CORD and the entire rare disease community, Rare Disease Day 2022 is an occasion for celebration, even as we continue the fight to beat back COVID-19, because it's the turning point for affordable and sustainable access to the most appropriate therapies for rare diseases. We're confident of success on both fronts. ■

To learn more about CORD's initiatives in the rare disease space, visit **raredisorders.ca**.

Read SMA patient Jared Wayland's story on pg 4.

Strategic Account Director: **Jessica Golyatov** Strategic Account Manager: **Madison Trajkovski** Country Manager: **Nina Theodorlis** Content & Production Manager: **Raymond Fan**
Designer: **Kylie Armishaw** Content & Web Editor: **Karthik Talwar** Content Strategist: **Emma Yordanov** All images are from Getty Images unless otherwise credited.
This section was created by Mediaplanet and did not involve Maclean's magazine or its editorial departments. Send all inquiries to ca.editorial@mediaplanet.com.

PHOTO COURTESY OF GHASSAN AKL

Ghassan Akl

Taking Advantage of Every Moment Life Has to Offer

Living with generalized myasthenia gravis used to mean leading a very restricted life. New options are now changing that.

Anne Papmehl

When Ghassan Akl was 17, he suddenly found himself unable to lift a glass of water. As a very active teenager who worked out at the gym every day, this was unusual. His parents brought him to the hospital where his symptoms were recognized, and after a series of tests his diagnosis was confirmed.

That diagnosis was generalized myasthenia gravis (gMG), an autoimmune neuromuscular disorder in which the immune system mistakenly produces antibodies that block receptors in the neuromuscular junction — the area where nerve cells connect with the muscles.

A gradual loss of independence

Hallmark signs and symptoms of this rare disease involve fluctuating weakness of the voluntary muscles. "Many different muscle groups may be implicated, resulting in various patterns of weakness," says Dr. Amanda Fiander, a neurologist at a private clinic called Maritime Neurology in Halifax, N.S. "In addition to arm or leg weakness, possible symptoms may include double or blurred vision, drooping eyelids, slurred speech, difficulty chewing or swallowing, and shortness of breath."

Akl had most of these classic gMG symptoms. Over the next 11 years, he became increasingly dependent on family members to help him with simple things like going up the stairs. During this time, he underwent conventional immunotherapy treatment, which involved going to the hospital twice a week from 8:30 a.m. to 3:00 p.m. Eventually, as the disease progressed and became more aggressive, he had to stop working and going to school.

Treatment for gMG has remained pretty much the same for the past 50 or 60 years. However, new treatment options are allowing for more effective disease management and an overall improvement in the quality of life for Canadians living with gMG. Since starting on one of them two years ago, Akl has become a different person. "I'm able to train at the gym, run, and be independent again," says Akl, now 30. He's currently training to become a barber and recently returned to school.

The importance of recognizing signs and symptoms

Luckily for Akl, because the emergency room nurse recognized gMG, he was put on the right diagnostic track and into treatment very quickly. "Recognition of the disease is essential as gMG can lead to significant disability," says Dr. Fiander. That can sometimes be tricky since some gMG symptoms may overlap with other neurological disorders or even non-neurologic medical conditions.

In addition to a correct diagnosis, it's critical that people living with gMG have regular assessments from their health care providers (HCPs) to monitor their disease, something that, according to his situation, Akl does once a year with his doctor. "Given the fluctuating nature of the disease, regular HCP assessments are important to ensure that the treatment approach is both effective and appropriate for the particular individual," says Dr. Fiander. "Disease symptoms may change over time — and can sometimes do so quickly, requiring a change or adjustment in medication. These adjustments are best made early on, to avoid crisis."

For Akl, life keeps getting better and better. No longer confined to spending more than 12 hours a week in the hospital, his new regimen requires only a half-hour hospital visit every two weeks. "I'm taking advantage of every moment I have," says Akl, who is now able to think about longer-term career goals. The medical field is one of the options he's considering. ■

Dr. Amanda Fiander
Neurologist,
Maritime Neurology

Talk to your doctor if you recognize the set of symptoms or to learn more visit **muscle.ca**

*This article was made possible with unrestricted support from **Alexion Pharma Canada.***

Why It's Important to Recognize Signs of Rare Blood Disease

The road to a PNH diagnosis can take up to five years. That's why awareness is critical for both patients and physicians.

Anne Papmehl

Paroxysmal nocturnal hemoglobinuria (PNH) is an ultra-rare and acquired blood disease caused by a mutation in some of the bone marrow stem cells. "The subsequent blood cells produced lose their protection against a part of our immune system called complement," says Dr. Christopher Patriquin, a hematologist and Clinician Investigator at the University Health Network and Assistant Professor of Medicine at the University of Toronto. This can lead to the destruction of the red blood cells, as well as damage, and activation of the platelets and white blood cells, which puts the patient at increased risk of thrombosis (blood clots). "Some patients with PNH may also have bone marrow failure, such as aplastic anemia, where blood cell production is significantly reduced," says Dr. Patriquin.

The median age for PNH diagnosis is between the ages of 35 and 40 — people in the prime of their career and family responsibilities. "It's devastating because when your red blood cells are being destroyed, you're not really able to function properly," says Cindy Anthony, Executive Director of the Aplastic Anemia and Myelodysplasia Association of Canada, a patient support organization.

The importance of recognizing signs and symptoms

"Undiagnosed, symptomatic PNH has a 35 percent five-year mortality rate with up to two thirds of deaths being due to thrombosis," says Dr. Loree Larratt, Professor Emeritus in Clinical Hematology at the University of Alberta. These severe blood clots can form virtually anywhere throughout the body, including the brain, heart, lungs, and kidneys. "Beyond the risk of thrombosis, untreated patients with PNH may develop many other complications from the continual breakdown of their red blood cells, such as fatigue, shortness of breath, abdominal pain, difficulty swallowing, and kidney damage," adds Dr. Patriquin.

Diagnosis is made by a test of the patient's blood called flow cytometry. But the challenge is recognizing that patients may have PNH to begin with and need to be tested. "It's important to recognize that the patient may present with some very common symptoms such as fatigue, anemia, or thrombosis, for which there are often other clinical explanations," says Dr. Patriquin.

Symptoms that may warrant testing include unexplained low blood counts, some types of bone marrow failure, thrombosis that may be unprovoked or presenting in an atypical location, red-coloured urine (called hemoglobinuria), and hemolytic anemia that is not otherwise explained.

> It's important to recognize that the patient may present with some very common symptoms such as fatigue, anemia, or thrombosis for which there are often other clinical explanations.

Hope for living well with PNH

While there's no cure for PNH other than a bone marrow transplant, which is usually reserved for severe cases of bone marrow failure, it's possible for people with PNH to live full and active lives. "Not all patients initially require therapy, but they should still be seen by a physician with expertise in PNH," says Dr. Larratt.

The core treatment strategy for PNH is to protect red blood cells from damage and destruction. Since 2009, a complement inhibitor has been approved in Canada, which is administered by intravenous infusion every other week. The medication effectively stops the hemolysis, thus improving the quality of life and increasing survival by reducing the risk for thrombosis for many PNH patients. "These new treatments have been a game-changer for sure," says Anthony.

Things are expected to get even better. "How we manage PNH 10 years from now will likely be quite different from how things are done today, with hopefully more options for our patients that continue to control the disease while also improving quality of life and reducing the treatment burden," says Dr. Patriquin. "That's why it's imperative that there is continued support in Canada for research into PNH and its treatment." ■

Dr. Chris Patriquin
Hematologist, Clinician Investigator, University Health Network & Assistant Professor of Medicine, University of Toronto

Cindy Anthony
Executive Director, Aplastic Anemia & Myelodysplasia Association of Canada (AAMAC)

Dr. Loree Larratt
MD & Professor Emeritus, Clinical Hematology, University of Alberta

Talk to your doctor if you recognize the symptoms or learn more by visiting **aamac.ca.**

*This article was made possible with unrestricted support from **Alexion Pharma Canada.***

New Father with Rare Disease Works Hard to Access Potentially Life-changing Treatment

When you live with a progressive neuromuscular disease like spinal muscular atrophy (SMA), timely access to treatment is critical to retaining strength and ability.

Anne Papmehl

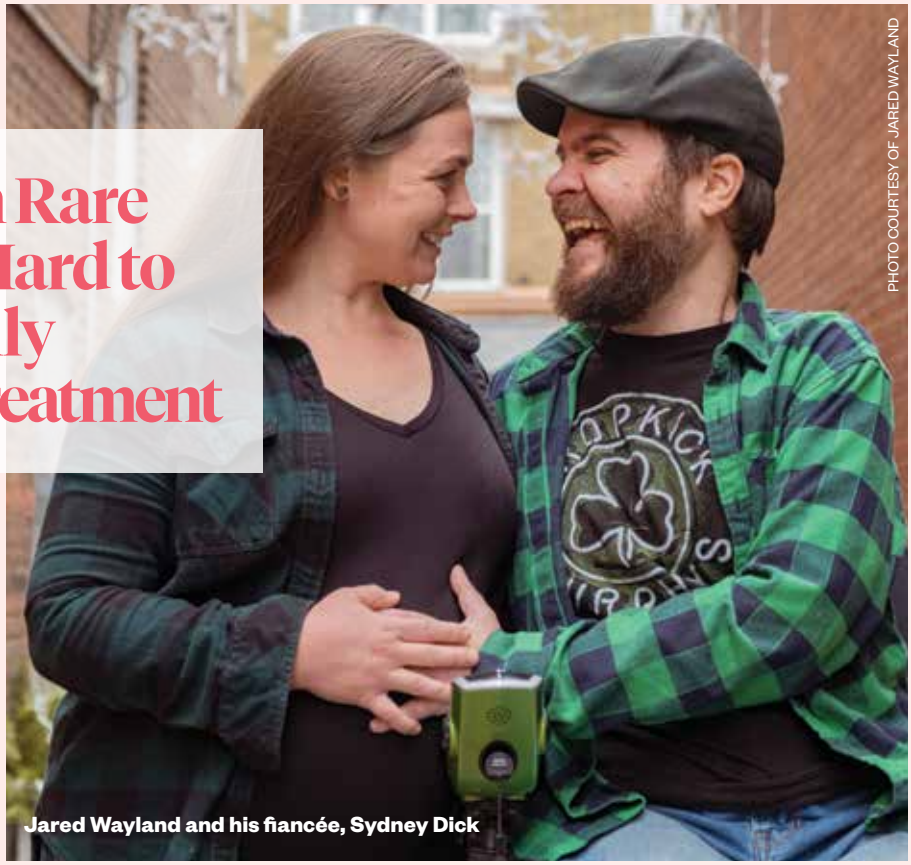


PHOTO COURTESY OF JARED WAYLAND

Jared Wayland and his fiancée, Sydney Dick

Jared Wayland and Sydney Dick of St. Catharines, Ont. have been looking forward to the birth of their first baby, who entered the world in early January. Their hopes and dreams are typical of many first-time parents — that their child grow up to be happy, healthy, and loved.

But Jared and Sydney are not typical parents. Jared, 33, lives with SMA, a rare, genetic neuromuscular disease that has already robbed him of his ability to walk and continues to make him weaker with each passing day. SMA is characterized by loss of motor neurons in the spinal cord and lower brain stem, resulting in severe, progressive muscle atrophy and functional decline.

"I'm determined to live my life to the fullest, but the future is always uncertain for me and my family because of my SMA," Jared says. "Sydney and I will have the same types of struggles as any other new parents, but sometimes I wonder, 'will I even have the strength to hold my child a few years from now?'"

Simple day-to-day tasks a huge challenge

Jared lives with Type 3 SMA, a later-onset form of the disease. Prior to his diagnosis 20 years ago he was an active teenager, participating in hockey, baseball, swimming, and golf. But Jared began to notice it was getting harder to get up after falling and he was tripping a lot. "One day I was holding something, and my parents noticed my hands were shaking," he says. A trip to the family doctor led to several neurologist visits and an eventual SMA diagnosis.

Although treatment for SMA was non-existent throughout his adolescence and early adulthood, therapies have since been approved in Canada. However, many adult patients, including Jared, are still waiting for access. With the exception of Quebec, where treatment is available for all adults, most provincial reimbursement guidelines permit access in exceptional situations, evaluated on a case-by-case basis.

An application for treatment reimbursement was made for Jared by his physician in August 2019. More than two years later it continues to be under evaluation, its outcome elusive and far from certain — a reality shared by the majority of adult patients outside Quebec who apply for access. Meanwhile precious time and muscle have been lost and Jared's condition continues to decline. He now relies on a power wheelchair almost exclusively for mobility and counts on Sydney to help him with much of his care, from getting up in the morning to bathing and dressing.

"Treating my SMA as soon as possible is critical to ensuring I can help care for my child and be the father I've always dreamed of being. We know it's a time-sensitive issue and that my SMA will continue to progress. Now with the baby here it only expedites the need," says Jared. "Just being able to maintain what I still have would be great, but if I could go back to the level of ability I had just two years ago, that would be a big plus."

Staying optimistic amid challenges and advocating for others

As Sydney's due date approached, her ability to physically support Jared became increasingly limited. To provide support to his family, Jared invested in costly lifts that increase his independence and help him fulfill his caregiving goals. With the baby now here, the couple will face similar struggles as all new parents do, as well as change their care dynamic as attention shifts more to their child's needs. "Fortunately, we have both sets of parents and siblings nearby to help, but we're still figuring how things will play out. Access to treatment for Jared would be one less thing for us to worry about," says Sydney.

Despite his challenges, Jared is optimistic and counts his blessings. He is excited to be a new father and says he is committed to raising his child in a home that sets an example for inclusiveness and understanding. He continues to have the use of his hands and is currently able to earn a successful living as a graphic designer. Over the past five years, Jared has also been cultivating a positive mindset and advocating for treatment access for all adults. "As important as this is to us, it's a much wider issue. There are so many people out there who could benefit from treatment, and anything I can do to help is what I want to do," he says. ■

If you or a loved one is living with SMA, please visit **facebook.com/smashingbarriers.ca**.

This article was made possible with support from a research-based pharmaceutical company.