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RARE DISEASES

PHOTO: DENNIS KUZMAR

UNITING FOR CHANGE

The Johnson family stands together to rally against Prader-Willi Syndrome and raise community awareness.

Featuring

FRESH HOPE FOR PATIENTS
Advancements in hemophilia treatment

NAVIGATING THE SYSTEM
Providing patient support

REMOVING BARRIERS
Improving access for equal treatment

Who needs a 'Canadian Rare Disease Strategy'?

Durhane Wong-Rieger
PRESIDENT & CEO,
CANADIAN ORGANIZATION
FOR RARE DISORDERS

For starters, the nearly 3 million Canadians living with a rare disease and their families:

Patients like Eric, who struggled for 30 years seeking a cause for his many painful and debilitating symptoms. It wasn't until his infant daughter developed the same symptoms that the doctors figured out they both had a genetic auto-inflammatory disorder. Or the Sean family, who live in a province that does not screen for Joey's rare metabolic disorder. His parents had two more boys with the same condition before they realized something was seriously wrong. Or Gerald, who thought he had won the lottery when he found a clinical trial for his son's rare condition. Then he learned that no trial sites were planned for in Canada. Perhaps Richard, who was told he would not get access to a drug for his progressive neuromuscular disorder until he became "more disabled." His brother, in the neighboring province is being treated.

What about Claire and Al, whose two-year-old daughter received a diagnosis of a rare progressive pituitary disorder? They were given a long list of tests and specialists she would need the rest of her life, an even longer list of physical, cognitive and behavioural challenges, and a very short list of supportive therapy, and no contact information for counselors, social workers or parent support groups.

A strategy to improve lives

In March 2015, at this year's Rare Disease Day Conference, the Canadian Organization for Rare Disorders released a 50-page report on "Rare Disease Strategy for All Canadians." The culmination of three-years of multi-stakeholder consultation, the document sets forth to improve the lives of patients and families in eight goals.

'The Strategy's' eight goals are:

1. Public awareness of rare diseases and their public health impact.
2. An environment that is responsive to the wide-ranging needs of widely diverse rare disease patients and families.
3. Prevention and early detection of rare diseases are regarded as important goals of public health.
4. Communities resourced to support individuals with rare diseases and their families.
5. Timely and equitable access to seamless care for all Canadians with rare diseases, regardless of where they live.
6. All decisions informed by the best available evidence generated throughout the course of disease.
7. Sustainable mechanisms for access to promising rare disease therapies.
8. A world leader in enabling and fostering innovative research around the prevention, diagnosis and management of rare diseases.

Each goal defines specific actions involving all stakeholders. Our top priority is directed to the Federal government to implement, without further delay, the Orphan Drug Regulatory Framework, announced by the Health Minister in October 2012, which will establish an official definition of rare diseases and stimulate research and development for new therapies, including clinical trials in Canada. Moreover, Canada needs a 'National Research Program for Rare Diseases' as well as a commitment to sustained funding. The medical colleges as well as at the professional associations need to develop and implement education and training in their curriculum and continuing education. 'The Strategy' identifies an urgent need to include next-generation genetic testing and genetic counseling (about 80 percent of rare disease are genetic).

Change through unity and collaboration

'The Strategy' also calls for linkages to education, employment, disability, and other community services. We need the private and public drug plans to work with the drug manufacturers to establish mechanisms that will assure appropriate, timely, and sustainable access to drugs as soon as they are approved. And we need investment in new drug development as well as support for the "repurposing" of existing drugs for rare diseases. Finally, patient groups must be appropriately resourced to partner in every area, including identifying unmet needs for drug discovery, designing clinical trials, establishing patient registries, developing patient-centred best practice guidelines and treatment protocols, developing and linking support services and care, and serving as a resource to patients and families.

So, who needs a 'Canadian Rare Disease Strategy'?

Just about everyone, including the payers and public health programs, healthcare and supportive care providers, researchers and drug developers, and patient groups and all Canadians.

This spring, CORD will host a regional and provincial consultation on the 'Rare Disease Strategy'. More than twenty-four European countries have approved national plans for rare disease, and our goal is to have all-stakeholder support for a Canadian strategy by fall 2015.

DURHANE WONG-RIEGER
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THE TRUE HARD FACTS: The journey of a rare disease patient

RECEIVING TREATMENT Young girl, Maya, with MPS I, receives treatment at home with brother Owen.
PHOTO: SUBMITTED

You have a child with chronic, often severe, symptoms that your family physician can't explain. Can you imagine seeing six or more specialists over a period of three to 11 years just to get a diagnosis?

That's what we heard from one-third of the 550 respondents to the first survey of Canadians living with rare diseases. The Canadian Organization of Rare Disorders conducted the survey between January and March of 2015. Moreover, one-third received more than three misdiagnoses, with one-tenth getting more than five wrong diagnoses.

The need for a comprehensive national plan

According to Durhane Wong-Rieger, President & CEO, "CORD did the survey because we knew Canadians with rare diseases were struggling with everything from getting a diagnosis to finding a specialist to accessing treatment, but we had no data. The French did a survey about 10 years ago, the Australians three years ago, and the Brits just last year, all underscoring the need for a comprehensive national plan for rare diseases. We were planning to present a 'Canadian Strategy for Rare Diseases' on Rare Disease Day this year, and we felt Canadian data would help make the case.

As challenging as it was to get an accurate diagnosis, the participants' experience of the information and support received at the time of diagnosis (and afterwards) is best described as "woefully inadequate" for more than half of the respondents. Nearly half did not get the right amount of information and only half felt they understood the information provided by the doctor. Most lacking was a follow-up, with more than two-thirds reporting they were given no individual or contact number for follow-up, nor were they referred to a patient organization.

Fighting for access

An overwhelming majority (four-fifths) of participants felt GPs were not informed about rare diseases, and more than half said neither the pediatricians nor the specialists were informed. More than two-thirds of the English participants and 90 percent of the French respondents felt that educational services were not aware of rare diseases, and similar perceptions were offered about places of employment and disability services. "Because these services are not aware of rare diseases, our children (we) do not get the same support or consideration as those with more well-known conditions like cancer, heart disease, and diabetes."

There are few rare disease clinics and even fewer with comprehensive care services. Not surprisingly, then, more than half feel they do not get access to specialists or clinics and there is little coordination of care.

About one-third said they could not access appropriate drug treatments, and the same number said they were not able to access "non-drug" treatments.

Provincial disparities

The survey also confirmed that patients and families incur significant personal costs in dealing with their rare disease. Those living outside of Quebec (English respondents) had more personal expenses than did those in Quebec. On average, non-Quebeckers paid almost $3,000 "out-of-pocket" for health services such as testing and physiotherapy, while Quebeckers paid less than $2,500 annually. In terms of drug costs, those in Quebec (French respondents) paid nearly $1,000 a year less than those in the rest of Canada ($1300 as compared to $2300). A similar differential is experienced for support services, average annual expenses of $1,430 for Quebeckers and $2,510 for all others. Finally, families outside of Quebec pay about 60 percent more for all of their other disease-related items than do Quebeckers, average expenses of $6,560 versus $3,814, respectively. Taking all categories into consideration, the average non-Quebec rare disease family is paying up to $14,400 every year out-of-pocket and about $9,000 annually, in Quebec.

In summary, based on feedback from the first ever direct survey of Canadians affected by rare diseases, most patients and families are not getting timely access to diagnosis, health services and community support services. There is a lack of an infrastructure for dealing with rare diseases as a public health issue, which stems from the lack of awareness among health professionals and the community at large.

The burden of illness

The costs to the healthcare system may be even greater than feared, when families are consulting up to 20 healthcare professionals over a period of six or more years, just to get to a diagnosis. Following that, there is no coordinated care. Most patients feel that they are on their own and continue to seek the right treatments and supports, often without a contact person for the information needed. True burden of illness studies are needed though specific studies in other countries have indicated that costs may be seven to 16 times higher for a rare disease as compared to more common ones, with correspondingly higher impact on families.

Certainly, in the Canadian survey, this latter appears to be substantiated, with families spending on average, more than $10,000 on disease-related costs not covered by the public or private health plans.

> "There is a lack of an infrastructure for dealing with rare diseases as a public health issue, which stems from the lack of awareness among health professionals and the community at large."

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EDITOR'S PICK

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Getting the conversation started: Let's raise awareness on tuberous sclerosis complex (TSC)

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WHAT IT'S LIKE TO LIVE WITH A RARE DISEASE IN CANADA:

A brief summary of the results from the first ever survey of Canadian patients and families living with rare diseases from 500+ participants across Canada, including Quebec, conducted by CORD.

ACCESS TO TREATMENT AND SUPPORT

Less than **40%** have access to a rare disease specialist or clinic

Less than **40%** have access to drug treatments

Less than **30%** have access to non-drug treatments

GETTING TO DIAGNOSIS

Median time to diagnosis: 3 to 6 years — **21%** over 6 years

Median number of specialists consulted: 3 to 5 — **10%** over 11 specialists; **5%** over 20 specialists

Median number of "wrong" diagnosis: 1 to 2 — **40%** more than 3 misdiagnoses; **15%** more than 5 misdiagnoses

INFORMATION AT DIAGNOSIS

ONLY 40% got the right amount of information at diagnosis

ONLY 50% understood the information provided by the healthcare professional

ONLY 50% know where to get the information needed

SOURCE: CORD PATIENT SURVEY 2015

Hemophilia: 50 years of progress

Advancements in the care and treatment of inherited bleeding disorders from the early 50's to present.

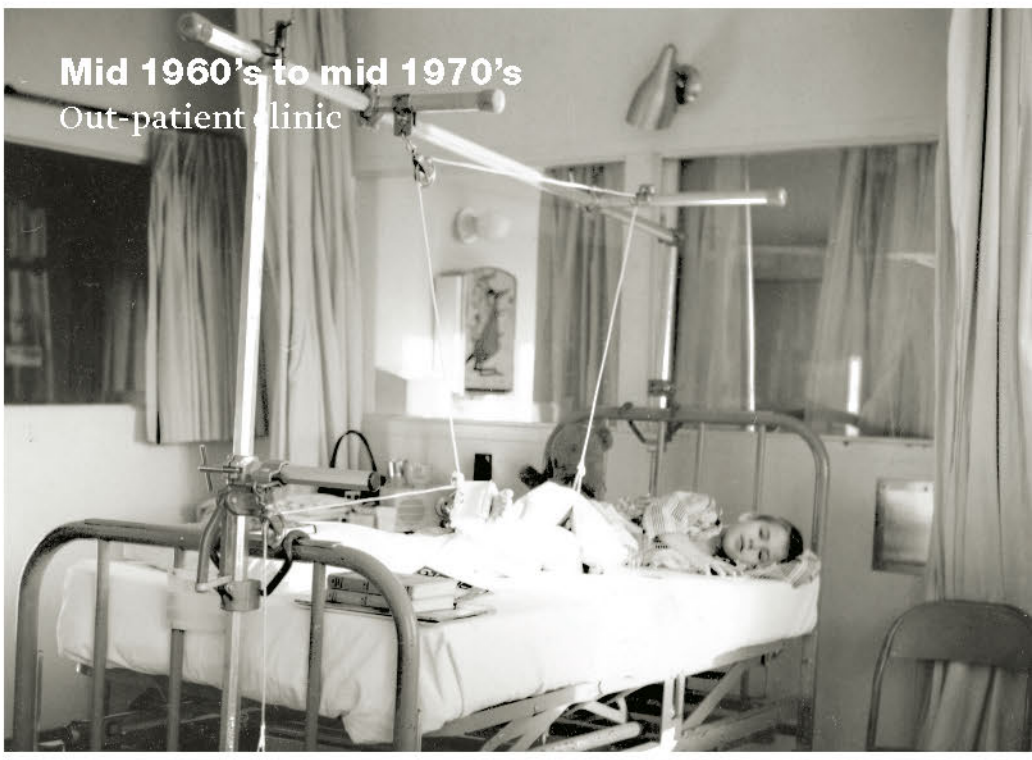
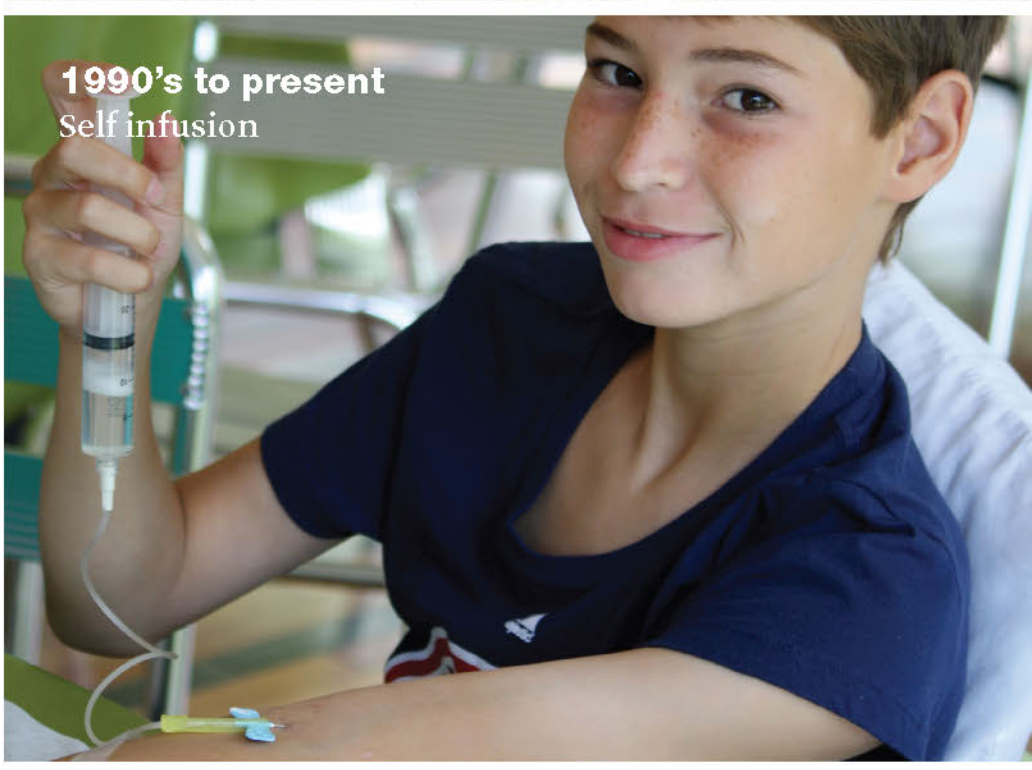


PHOTO: THE CANADIAN HEMOPHILIA SOCIETY



In 1965, Dr. Judith Pool of Stanford University discovered that the sludge on the surface of human plasma after centrifugation was rich in factor VIII, the protein missing in the blood of people with hemophilia A. Skimmed off and transfused, it proved far more effective in stopping bleeding than whole blood or plasma, and revolutionized treatment.

People with hemophilia A, and its rarer cousin hemophilia B (a deficiency in factor IX), suffer from internal bleeding, mainly into joints and muscles, and occasionally into vital organs, which can be fatal. Before Dr. Pool's breakthrough, life expectancy for people with hemophilia was less than 20 years, and their lives were full of the excruciating pain of internal bleeding. Repeated hemorrhages into knees, ankles and elbows led to severe crippling.

> "Today, 50 years after the discovery of cryoprecipitate, more advances are on the horizon. Extended half-life factor concentrates are being introduced in Canada."

Improving quality of life

The discovery of cryoprecipitate quickly led to a further advance, the development of plasma-derived factor VIII and IX clotting factor concentrates, lyophilized powders that could be refrigerated for long periods, reconstituted and infused intravenously by the patient himself at home or anywhere in the world. In the 1970's, a network of 25 hemophilia treatment centres sprang up across Canada to support people with hemophilia, based on the concepts of interdisciplinary care and home infusion. This meant freedom from hospital admissions and emergency room visits, and more normal lives!

It seemed too good to be true... and it was. Factor concentrates were made from the pooled plasma donations of tens of thousands of donors; no technology existed to eliminate viruses from the products. They transmitted hepatitis C and often, HIV. It was a huge public health tragedy.

Prioritizing safety

Massive efforts were made to make these concentrates safe. In the mid and late-1980's, very effective viral reduction methods were introduced to treat blood products. There has not been a single case of HIV or hepatitis C transmission with hemophilia products in Canada since 1988. Then in the early 1990's, came the introduction of recombinant factor concentrates, in which the human gene to produce factor VIII or IX is placed in animal cells and grown without the use of human blood. Prophylaxis, the regular infusion of factor VIII or IX to prevent bleeding, gradually replaced the older approach of infusing after the bleeding—and the damage—had occurred.

Today, 50 years after the discovery of cryoprecipitate, more advances are on the horizon. Extended half-life factor concentrates are being introduced in Canada. These offer the promise of less frequent infusions, more effective prevention of bleeding and higher quality of life. Gene therapy for hemophilia B is in early clinical trials and, if proven safe and effective, would mean an end to the one to seven weekly intravenous infusions currently required to treat hemophilia effectively.

A hopeful future

There remain important challenges. The immune systems of a small but significant percentage of people with hemophilia reject factor concentrates. This inhibitor rapidly destroys the factor VIII or IX as soon as it is infused. Therapy is much less effective. While modern treatments are freely available to people in countries like Canada, this is sadly not the case worldwide, where three-quarters of people with hemophilia receive little or no care at all.

Given the progress of the last 50 years, however, there is every reason to be hopeful.

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INSIGHT

Major complication of hemophilia treatment warrants **new approach**

NEW APPROACH GIVES NEW POSSIBILITIES
Nathan (left), who has severe hemophilia A, enjoys many activities and lives life just like other children with a few adjustments.
PHOTO: SUBMITTED

New approaches to hemophilia A treatment are giving renewed hope to sufferers of the condition who have inhibitors to the standard treatment, factor VIII (FVIII) or those at high risk of developing inhibitors.

Hemophilia is a rare, genetic lifelong condition in which a patient's blood lacks the required blood-clotting factors and, therefore, cannot clot normally. Whereas, after a receiving a cut or injury, a regular person will stop bleeding relatively quickly, somebody with hemophilia will bleed for a much longer period of time. This can make any small injury potentially hazardous, and can become a major health issue if a hemophilia patient experiences internal bleeding and cause long term damage.

Significant complication

In most cases, hemophilia A patients can be treated with replacement infusions of FVIII, the clotting factor that hemophilia A patients lack. This factor can either be derived from purified human plasma or made using recombinant technology. However, some hemophilia A patients develop antibodies (inhibitors) that reduce the clotting action of the FVIII product and make its infusions either less effective or completely ineffective. Sometimes very high doses of FVIII concentrate are administered to try to override the antibody to FVIII that the body has made.

"From the medical point of view, inhibitors to FVIII are the most significant complication arising from the treatment in patients with hemophilia," says Dr. Anthony Chan, Professor, Pediatrics, Director of the Hemophilia Program at McMaster Children's Hospital. "If a patient is bleeding into a joint, long term joint damage can be caused. Patients can also have life threatening bleeding."

Wide scale impacts of FVIII inhibitors

As well as the medical impacts, patients with inhibitors to FVIII can often find that every aspect of their life is affected by not having access to effective blood clotting medication. "It can cause concern in all daily activities because there is no way to plan for bleeding, it's spontaneous," says Professor Chan. "It can also cause problems for a patient who plans to travel for work, or children participating in their daily activities, but will have worries about bleeding while they're away from their own treatment centre, where the doctors know them very well."

> "The way that he's able to live his life really speaks to the quality of the treatment."

Inhibitors to FVIII also increase the strain and create practical problems for the parents or caregivers of hemophilia patients. There is a wider societal impact too, as significantly more medical resources are required to treat a patient with inhibitors to FVIII compared with a regular hemophilia patient. "Compared with hemophilia patients without inhibitor, physicians and the multidisciplinary team spend a lot more of their time dealing with patients with inhibitors, devising strategy and monitoring them a lot more closely," says Professor Chan.

A story of hope

Although life for patients with inhibitors to FVIII can be more complicated, the story of Nathan, who was diagnosed with severe hemophilia A at birth, can give fresh hope to others affected.

Just five weeks after Nathan's birth, his brother, Benjamin - who also has hemophilia A, developed an inhibitor after being treated with recombinant factor VIII for a severe tongue bleed. After an insertion of a device allowing frequent injections, Benjamin started a high dose factor VIII therapy to try to override the antibody to factor VIII and responded well, he's now inhibitor free.

Yet, the family's hemophilia team was understandably concerned that Nathan might too be susceptible to the development of an inhibitor, and therefore suggested that Nathan try an alternative type of factor that's produced from the plasma of human donors.

"We now treat Nathan prophylactically every other day with the plasma based factor, and since starting this treatment regime he has not had a bleed in over four years," said Nathan's parents, Victoria and Darryl.

Just like other kids, Nathan and Benjamin are able to enjoy the activities that make childhood a magical time. "For the most part, Nathan is able to live like any other six-year-old," says Darryl. "The way that he's able to live his life really speaks to the quality of the treatment."

In addition to ways to better manage inhibitors, research is also being conducted on ways to reduce inhibitor development. A new recombinant manufacturing technology has recently allowed development of a FVIII treatment that closely resembles the type of factor VIII produced in healthy humans, but without any exposure to human blood and without modifications to alter the way the body treats it.

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IMPROVING THE PROGNOSIS FOR TTP PATIENTS

A WALK IN HER SHOES
Every year, Sydney walks with her team to raise awareness and funds for TTP.
PHOTO: ANSWERING T.T.P FOUNDATION

In September of 2008 I got out of the pool after a swim, took off my goggles and suddenly couldn't see anything in front of me. Everything was dark. I was twenty-eight years-old, a new MBA graduate and recently married. I waited five hours in a major Toronto Emergency Room, only to be sent home with a migraine diagnosis. The next day the migraine was still there and my vision was fluctuating—but my family doctor's office told me there was nothing to worry about. My mom knew something wasn't right and sought a third opinion. Routine blood work revealed dangerous anomalies that resulted in an emergency referral to another hospital, and finally a diagnosis of Thrombotic Thrombocytopenic Purpura (TTP).

TTP is a rare blood disorder that is difficult to diagnose, but is considered a true medical emergency. TTP is estimated to affect 3 in 1 million people per year—mostly young women like me. It causes blood clots that can damage organs such as the brain, heart, kidneys. If not properly diagnosed and treated, mortality is as high as 95 percent.

Improving treatment

My treatment included daily replacement of my blood plasma (a component of blood), with that from healthy individuals using a process called plasmapheresis. When I was hooked up to the plasmapheresis machine, I felt like my whole body was humming. Reactions to the donor plasma are common and I was no exception; I was quickly treated with intravenous antihistamines to mitigate the chance of a full-blown anaphylaxis reaction.

About 60 percent of TTP patients will go through this treatment process once and never experience a relapse. For others, like me, TTP continues to reoccur. I have had six relapses and 68 plasmapheresis treatments so far. Over the past six years, my treatment has improved; my hospital has obtained updated equipment and I have qualified to receive an alternative donor plasma product called 'Solvent Detergent Plasma'. Compared to fresh frozen plasma, this product has been processed to remove certain blood born viruses and allergic reaction debris. It is frustrating to me that not all patients across Canada have access to this safer blood product.

Battling any disease is challenging; having a rare disease adds the element of feeling alone at times. I am thankful to have the constant support of my husband, family and close friends, many of whom have volunteered their time to help me establish the Answering T.T.P Foundation. This charity is the best thing that's come out of my TTP journey. Like most rare disease charities, skilled volunteers are our most precious resource. It is tough to find those willing to join us in the pursuit of improving the prognosis for all those affected by TTP.

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INSPIRATION

Mississauga parents rally around son, fighting for rare disease community

Keegan and Tanya Johnson knew something wasn't right with their newborn son. Dante could barely move and he never cried. He had little to no suck reflex. When tests revealed he had a rare genetic disorder called Prader-Willi Syndrome (PWS), they were distraught.

But the Mississauga couple decided to take action. They educated themselves about PWS and soon reached out to people who, like them, faced the challenge of raising a child with cognitive disabilities, problem behaviour and a chronic feeling of hunger.

When Dante was just a few months old, they organized 'One Small Step', a walk that raised money for PWS research. Now, ten years later, there are 75 such walks worldwide. Combined, they have raised more than $5 million for PWS research.

In the past two years, the couple's focus has broadened to include other rare disorders— generally defined as those that affect fewer than five in 10,000 people — and they are active within this larger community.

"When we attended an event put on by a rare disease patient advocacy organization in the U.S., we met families whose children had fatal diseases. It was heartrending," Tanya says, noting that 30 percent of children with rare diseases die before the age of five. The couple later attended a Canadian Organization for Rare Disorders (CORD) conference and were inspired by individuals who are "pushing the [rare disease] community forward."

It occurred to the Johnsons that the people active in these various communities — from Cystic Fibrosis to Tay Sachs Disease and Progeria — could accomplish a lot by working together.

The couple organized 'Give Rare', a web-based event in which non-profit organizations for rare diseases competed against each other in fundraising. The event, which was held a few weeks ago, focused on U.S. organizations. It will come to Canada next year.

THE JOHNSON FAMILY
The Johnson family (top) and Dante (bottom) empower people to make a difference in their own communities.
PHOTOS: SUBMITTED

Parents send message about facing challenges

In all their endeavours, Keegan and Tanya are driven by deep love for Dante. Now 10 years old, he is like most boys his age in some respects. He is a happy kid who is passionate about sports. He is so caught up in 'March Madness', the annual NCAA college basketball tournament, he joined a pool.

But Dante has a difficult time managing his emotions — even a change in snack time can trigger a meltdown — and he has trouble learning. A special education teacher sits in on his classes.

One of the most troubling symptoms of PWS is an insatiable appetite. Without proper treatment, food becomes the central focus in the lives of people with the disorder and they often suffer from obesity. Fridge doors have to be locked and, in the case of adults, front doors do too — to prevent them from seeking food elsewhere. Some adults with PWS are forced to live in group homes.

To keep his appetite in check, Dante follows a modified ketogenic diet. He also takes growth hormones to increase his lean body mass. It was developed as a treatment for PWS less than 20 years ago.

The more advancements that are made in research and treatment, the more normal a life Dante and others like him can lead. That is the main reason Keegan and Tanya are working to heighten awareness of rare diseases, raise money for research and get government bodies on board with promising treatments. There is another motivation, too. "Both our kids are going to face challenges in their lives," says Keegan, referring to Dante and his younger brother, Denzel. "We want to send them the message that when you have a problem, you go out and fix it."

The Johnsons are doing all they can to tackle the problem of rare disease and they are hoping others will help.

"If you approached someone who has a family member suffering from a rare disease and asked him if he would contribute $500 to get some answers, the reply would be 'Yes,'" Keegan explains. "If the 30 million Americans affected by rare disease through their families and friends each gave $500 that would raise $15 billion. The equivalent figure in Canada would be $1.5 billion."

Where there is a will, there is a way.

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Giving a voice to Canadians affected by pituitary tumours

A national support network is playing a key role in improving the lives of Canadians affected by pituitary tumours.

The pituitary is a hormone-producing gland that is located at the base of skull. It has control over some crucial brain functions, including growth and maturity, fertility and sexual function, water balance, energy balance and response to stress.

Pituitary tumours are mostly benign and are separated into two categories: non-functioning tumours, which do not produce any hormones, and functioning tumours, which produce one or more pituitary hormones.

Acromegaly and Cushing's

"The two most common conditions caused by functioning pituitary tumours are acromegaly and Cushing's," explains Durhane Wong-Rieger, President and CEO of the Canadian Organization for Rare Disorders (CORD). "They are highly varied in terms of symptoms, but particularly challenging in that both conditions are changes in physical appearance."

Acromegaly occurs when the pituitary gland produces too much growth hormone. Common symptoms include excess growth of the hands and feet, changes in facial appearance including thickened lips and nose, a thickening of the scalp, and the jaw becoming more prominent. If possible, the preferred treatment option is surgery to remove the tumour. If it's not possible to remove the entire tumour, there are medications that may be used to reduce the amount of growth hormones to a normal level.

Cushing's is caused by the production of adrenocorticotropic hormone (ACTH), and symptoms can include weight gain, facial puffiness, diabetes, fatigue, high blood pressure and muscle weakness.

Creating The Canadian Pituitary Tumour Network

Symptoms associated with acromegaly and Cushing's often occur later in life and without warning. Because symptoms may develop slowly over time, patients may have a long period of "not feeling well" before they are diagnosed, often when complications have become severe. Many patients report feeling isolated and withdrawing from social activities, partly because of changes in physical appearance.

In certain parts of the country, patient support groups have been formed to provide information, emotional support and coping strategies. "Support groups play a tremendously important role," Wong-Rieger says. "Simply by being with others who share what you are going through is reassuring. As importantly, you have role models living with pituitary tumours who demonstrate acceptance, hope, and inspiration."

Inspired by the support groups, the Canadian Organization for Rare Disorders (CORD) has helped launch a Canadian Pituitary Tumour Network, which aims to bring together all those affected by pituitary tumours.

"As well as providing support for patients, we want to raise awareness among the public and the professionals, to encourage those affected to reach out and speak up and to promote development of diagnostic and treatment guidelines, so that patients are more readily diagnosed and have access to a network of informed practitioners," Wong-Rieger says.

Improving patients' access to effective treatments is another important role that the national network will play. "Because there are so few therapies specific to acromegaly and Cushing's, many current therapies are prescribed "off-label" and even those that are approved specifically for Cushing's or acromegaly are often not available universally," says Wong-Rieger. "In some cases, even private drug plans won't cover these medications. The Canadian Pituitary Tumour Network will help to ensure that there are good standards and guidelines for access to these therapies."

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INSIGHT

Getting the conversation started:

Let's raise awareness on tuberous sclerosis complex (TSC)

Raising public awareness of tuberous sclerosis complex (TSC) could play a major role in improving the prognosis of those affected by the disease and relieving the strain experienced by family members and caregivers.

Neurological disorder

Affecting approximately 3,500 Canadians, TSC is a neurological, genetic disorder that causes tumours and lesions to develop in multiple organ systems, including the brain, heart, kidneys, skin, eyes and lungs. In one third of cases, TSC is inherited from a parent, but, in the other two thirds, the disease is sporadic, occurring in a family's genetics for the first time.

"Manifestations vary widely, from mild to catastrophic," says Cathy Evanochko, whose daughter, Kimberly, was diagnosed with TSC aged 13 months. "Serious complications include epilepsy, kidney failure, heart arrhythmia, lung failure, cognitive disability and facial disfiguration; symptoms develop over the course of a lifetime."

Creating a support network

Cathy is a Co-Chair of Tuberous Sclerosis Canada SclérosTubéreuse (TSCST); a patient organization which aims to support families affected and share knowledge and understanding about diagnosis, management of the condition, and treatment options. When Kimberly was first diagnosed, all that Cathy had to refer to was a hospital pamphlet. Cathy wants more for families affected nowadays, she wants everybody to have easy access to all of the relevant information and guidance.

"As an organization, we want to make sure that Canadian families are in a position to take advantage of the best possible new treatments, which, at the moment, are coming along very quickly," Cathy explains.

> "Doctors have to be working together; we can't just have a neurologist working in one hospital and a nephrologist working in another. This approach is key to successful treatment of TSC."

A multi-disciplinary approach

New multi-disciplinary programs and clinics are specifically tailoring care for patients with TSC. "The model has come out of collaborations with our TSC international partners," says Cathy. "With a disease like this, doctors have to be working together; we can't just have a neurologist working in one hospital and a nephrologist working in another. This approach is key to successful treatment of TSC."

The introduction of the international 'Consensus Guidelines for Diagnosis and Treatment of TSC' has been vital in ensuring that individuals living with the condition receive consistent care, regardless of where they live or what hospital that they attend.

"Better understanding of TSC is key to unlocking cures and treatments for many conditions," says Cathy. "The life journey of patients and families is still not easy, but with research and heightened awareness it is becoming a road less rocky. As new treatments become available, the journey becomes one of possibilities and hope."

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KIMBERLY EVANOCHKO
Kimberly was diagnosed with TSC at thirteen months old.
PHOTO: CATHY EVANOCHKO

Dealing with the sticker shock of a rare disease

ACCESSING SUPPORT
Pharmaceutical companies offer various resources to help patients access and understand their personalized treatment options.

A recent CORD survey found that the median time to have a rare disease diagnosed is three to six years. If you are lucky enough to have a treatment for your condition, you may find yourself in sticker shock. Prescription drugs for rare diseases can cost in excess of hundreds of thousands of dollars or more per patient per year. In the same survey, over 70 percent of the patients indicated they had to incur personal costs for their treatment.

"When a patient gets diagnosed with a rare or orphan disease they don't understand all the hoops you have to jump through to access these drugs," said Sandra Anderson, a VP of Consulting and Business Development at Innomar Strategies which designs and operates Patient Support Programs.

Patients cannot simply go to the pharmacy to pick up a $100,000 drug, because they don't keep them on the shelf.

End-to-end support

Pharmaceutical companies offer 'Patient Support Programs' that help patients navigate through accessing coverage and reimbursement from private insurers and public health programs, and provide many different types of logistics and education support along the way. 'Patient Support Programs' help patients ensure that they can get on the needed medication as quickly as possible.

When a patient, or their child, is prescribed a specialty drug for a rare disease, their doctor will typically connect them to that drug's 'Patient Support Program', starting with a phone number, fax or online registration.

Financial guidance

When a drug costs hundreds of thousand dollars yearly, even the deductible can be prohibitive. With the lack of funding and intensive review process for these drugs, patients cannot always wait to get these drugs funded by the payers. The 'Patient Support Program' provider can help patients with the paperwork, authorization forms and the back-and-forth between insurers, public coverage and their physicians. In many cases they also seek financial assistance from the manufacturer which can offset the deductible that patients may be required to pay.

If you move, change or lose your job, 'Patient Support Programs' can pave the way with your new insurers, and try to ensure you have ongoing and uninterrupted access to your therapy, for example, if you need to find a clinic near your new home to administer your treatment.

Nurse and dietician support

These programs can offer infusion or injection services when required, and also dietician and nurse support to educate patients on how to follow the regimen of your therapy. They also support patients with the difficulties and challenges of living with a rare disease. Every program is different, customized to the drug, and the needs of the patient and their families. The programs also support the already established support network of physicians and clinics in the community and act as a partner to the limited support in place in the community. This is critical given that only 40 percent of the patients in the CORD survey felt they received enough information at their initial diagnosis.

Being diagnosed with a rare disease is often a shock in itself, and navigating through the system and facing the possibility of having to pay for the one drug that can help you can be overwhelming. Pharmaceutical manufacturers invest in 'Patient Support Programs' to ensure patients can get access to the medications quickly, affordably and are supported with the right information to help ease their journey.

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NEWS

Canadians struggle to gain access to life-saving drugs

TOP-TIER HEALTHCARE FOR ALL
Canada is introducing a new regulatory pathway for orphan drugs.

Rare diseases, collectively, affect millions of Canadians. These diseases can affect every system in the body from the cardiovascular, to the neurological, to the autoimmune.

Some, like relapsing polychondritis, affect only a few people per million; others, like cystic fibrosis, affect many more. Many of these diseases are life threatening, and about half of them target children and infants. And yet, for those living with a rare disease in Canada, getting prompt and affordable access to the treatments that can save or improve their lives can be a serious challenge.

The fewer customers, the larger the price tag

Rare diseases are often called orphan diseases because there is proportionally less research committed to their treatment or cure, leaving them stranded. And when a pharmaceutical company does develop a drug that is effective for a disease that may only affect a hundred people, they must charge much more per unit in order to recoup their investment.

For those Canadians with a rare disease and no drug plan, this can be devastating. And even for those who do get reimbursement from their insurance provider, it can present some serious financial problems. "Some plans have co-pays that can be expressed as a percentage of the cost," explains Stephen Frank, VP Policy Development and Health at the Canadian Life Health Insurance Association. "But a small percentage of a very large cost can still be very challenging for people."

Further, drugs for rare diseases (or orphan drugs) are often more difficult and expensive to get approved and bring to market than other drugs, in part due to having fewer potential subjects for clinical trials. This is an area where Canada's track record has been poor in the past. In fact, only 60 percent of treatments for rare disorders are ever approved in Canada, often years after they have been available in the United States and Europe.

A new framework for orphan drugs in Canada

Fortunately, things are changing. Canada is introducing a new regulatory pathway for orphan drugs that will provide an iterative approval process to allow companies to bring treatments for rare diseases to market while continuing to collect clinical evidence. "We're coming into a new world here with how orphan drugs are being brought to market in Canada," says Frank, "but that's only one piece of providing access. The other piece is how people are going to be able to pay for them, and that's still an evolving question both for public and private plans."

In the private sector, insurance companies have been banding together to create a pooling plan for extremely expensive rare disease treatments, in which all drug insurance carriers share the cost of whenever a customer needs the treatment. "The bigger the pool of people sharing the cost, the more sustainable these things are," says Frank. "Ultimately, though, in Canada we're going to have to get the public and private sector working together to find ways to address the cost and provide access."

And, by that definition, Canadians living with rare diseases are better insured than ever before. But the work is far from over. For the millions of Canadians in need of better access to, and reimbursement for, orphan drugs, we must continue to reinvest in the ideal of Canada's great health care system: top-tier health care for every Canadian.

And that's a job for all of us. "I don't think the private sector or the government will be able to solve this problem on their own," says Frank. "We're going to have to work together."

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CANADIAN SCIENCE BRIDGES OPPORTUNITY TO REALITY FOR TREATMENT OF RARE DISEASES

In recent years, great strides in research have been achieved in the field of rare diseases in Canada and internationally. In particular, a number of Canadian biopharmaceutical companies and institutional researchers have developed drugs for rare diseases that are saving and improving patients' lives. Many others are at the cutting edge of genetics research and personalized medicine, and are bringing forward the promise of life-saving medicines for Canadians impacted by rare disorders with no current therapies for their condition.

Many primary innovators and developers of biologic medicines used in treating rare diseases are working together to ensure creation of a regulatory pathway for rare disease medicines. With dozens of small and larger companies focusing research on debilitating rare diseases, Canada will join the community of global leaders seeking to improve the lives of patients.

Biotechnology offers hope

Orphan drugs are used in the treatment of rare disorders. These rare diseases affect less than 1 in 2000 people in Canada and are often severe, life threatening, and progressively debilitating genetic diseases. Biotechnology companies across Canada are developing medicines with the ability to offer hope to Canadian patients living with rare disorders. Canadian researchers and innovators are encouraged by the recent announcement to create accessibility for these medicines.

Patients living with a rare disease overcome hurdles every day. Important research in developing treatments for rare diseases is being done here in Canada, in company laboratories and throughout the research community. The ability to bring those treatments to patients in Canada is an objective shared by the government and industry.

With the intent to create a pathway for orphan medicines Canadian patients will now be on equal footing with other G20 countries. The initiative announced will help Canada to compete in attracting investment to nurture products into the marketplace, and see new solutions for unmet medical needs developed in Canada. The industry is encouraged by this initiative and looks forward to working with the Government to develop the final policy framework.

The time is now

There are numerous Canadian companies who have received orphan product designations for their products in various international jurisdictions. These Canadian solutions are offering hope to patients globally. With the initiative to create a regulatory pathway for rare disease medicines in Canada we are well on our way to offering the same solutions here at home. The U.S. Congress passed the Orphan Drug Act of 1983 to stimulate the development of medicines for rare diseases. Japan introduced similar legislation in 1993, Australia in 1998 and the EU in 2000.

It is time for Canada to recognize the opportunity to address the gap in care for patients with rare diseases.

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PERSONAL EXPENSES RELATED TO RARE DISEASE
(ON AVERAGE, EXCLUDING QUEBEC)

Personal expenses are disease related costs not covered by public or private health plans

Personal costs are about

60%

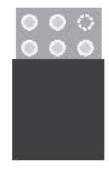
more for non-Quebeckers
than for Quebeckers

70%

incur personal costs related to rare disease

Average personal health services cost:
$3,000

Average personal prescription drug costs:
$2,300



Average drug costs OTC:
$800

Average support services costs:
$2,500

Average other costs:
$6,500

Average total personal costs:
Outside Quebec = $14,400, Quebec = $9,000

SOURCE: CORD Patient Survey 2015

SPOTLIGHT

Isabel Jordan
BOARD OF DIRECTORS, RARE DISEASE FOUNDATION

The "rare" intersection between online and offline networks

February 28th marked 'International Rare Disease Day' - an opportunity to bring greater awareness within and outside the rare disease community.

Several years ago some very smart people realized that while individual rare disease communities may be small, collectively the rare disease community is actually very large. Up to 1 in 12 people will have a rare disease in their lifetime, and the people facing a rare disease share a common journey with common obstacles, regardless of their specific disease. 'Rare Disease Day' is a chance to ignite the rare disease community to work together and achieve common goals as well as to increase the strength of our voice to those who wield power and make decisions.

A community effort

This year's 'Rare Disease Day' theme, 'Day-by-day, Hand-in-hand' started me thinking about how my offline and online world has influenced my experience as a parent of a child with a rare disease. My son, at 13, is undiagnosed. When we started his diagnostic journey we had no peers to turn to for answers or guidance. Creating our own rare community let me share experiences, find common goals, strategies and a voice for advocacy that gave meaning to our family's struggles.

Eventually our journey crossed into my social media world as it became more than just a place to share my kids' photos and lunch ideas. My network started to grow, giving me access to new perspectives and ideas. My "rare" social media community was becoming as important to me as my in person community, but in a different way. I became interested in how my online and offline communities might complement one another.

Making connections

How have my online networks changed my lived experience with rare disease? As a parent of a child with a rare disease I'm constantly looking for patterns, clues or ideas of what could be next in our diagnostic journey. I read postings by researchers, doctors and other parents. Could what they are saying have relevance to us? Can their approaches work for us?

In fact it was through reading someone else's blog that I began to see a pattern in my son's symptoms. Connecting the dots from someone else's story has allowed me to provide valuable clues to our own clinical team, and now we're heading down a new diagnostic path. Would I have seen them anyway? I don't know, but I credit my social media connections for helping me keep my eyes open for new ideas.

The rare disease community depends on social media. Statistically, we are spread thin across individual disease type. But together we can talk, connect, crowdsource ideas, and strategies. But I also think we can do it better. The question is, how?

Uniting across platforms

The answers may lie in finding the intersections between our online and offline networks. Lessons learned in my online networks have certainly informed my online connections and vice-versa. Can our organizations learn from this as well? Online, we are all equals and offline it can be easier to find empathy. These lessons of connection can bring us together.

A version of this piece was first posted on cyhealthcommunications.wordpress.com

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DARE TO THRIVE

STRONGER TOGETHER
Simon Ibell and long time friend, Steve Nash, work together to fight for funding for the weekly enzyme replacement therapy for MPS II (Hunter syndrome) in Ontario.
PHOTO: CAMERON GILBERT

Nine times in his life, Simon Ibell was told he wouldn't live to see the next year.

The first was when he was nearly two years old. His mother, Marie Ibell, had noticed his gait was off. At first, the pediatricians sent them home. They went on a global odyssey for a diagnosis and got one: Hunter disease or mucopolysaccharidosis II (MPS II), an ultra-rare genetic condition.

"They told my mom I wouldn't live to see the next year...that was in 1979 and now we're here in 2015," says Ibell.

A rare disease advocate

At age 37, Ibell is a renowned speaker, visionary and advocate for his own disease, and the rare diseases of others. In 2010, he founded the iBellieve Foundation, which strives to find a cure for MPS II and 50 related disorders.

"My disease has never defined me," he says. In 2002, he graduated from the University of Victoria with a degree in Sports Administration. When he was 24, he cycled 500 kilometres on Vancouver Island, raising $250,000 for MPS research in just ten days.

"I just got on with it," he says. "My attitude was very determined. This is what I have to deal with, and I just got on with it. It served me really well."

In 2003, he jumped at the opportunity of a lifetime, uprooting his Victoria life to take part in a clinical trial at the University of North Carolina Medical Center. He moved to Toronto so he could commute to North Carolina. That treatment ultimately stabilized his condition.

> "They told my mom I wouldn't live to see the next year...that was in 1979 and now we're here in 2015."

Recycling at a cellular level

Ibell explains his disease like this: "If you didn't have a way to recycle or get rid of your garbage in your kitchen that what's happening in every system of an MPS II patient's body."

There are 11 different types of MPS, says Jamie Myrah, the Executive Director of the Canadian MPS Society, but all of them lack the enzymes necessary for normal cell recycling and degradation, causing substances to build up in cells throughout the body.

She advises parents to watch for a "clustering of odd symptoms" in their child such as corneal clouding, frequent ear, nose and throat infections, joint stiffness, cardiac abnormalities and airway obstructions. Bone deformities and delayed growth are also common.

MPS patients face barriers where their prohibitively expensive treatment is not funded by the government or insurance providers, she says.

"But there's a lot of courage and hope out there. It's amazing to see what has happened over the last 10 years. The science is there, the technology is there now to really cure diseases like this, and these children grow up to live normal productive lives," she says.

The rare dare

For Ibell, advances in treatment enabled him to continue to make a difference in the lives of others.

In February, his organization launched the 'Dare 2 Be Rare' social media campaign, which encourages people to invite friends to take a "rare dare" and to raise or donate money if their friend does the dare. The campaign is raising funds for rare disease organizations, including his own, and highlighting the issues around rare diseases.

"The ironic thing with rare diseases is that individually they're rare. Collectively, they're not rare at all," he says.

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