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MEDIA PLANET

October 2012

# RARE DISEASES

3 FACTS YOU DIDN'T KNOW ABOUT RARE DISEASES

## COMING TOGETHER TO FACE THE CHALLENGES OF RARE DISEASES

One dozen Canadians with rare diseases made a journey to the Artic Circle **in support of research and awareness.**

PHOTO: SEBASTIEN LAROSE

# Canadian science brings patients hope

On October 3rd, the federal government announced that it will undertake the establishment of a national orphan drug strategy to ensure that Canadians with rare diseases have access to new orphan drug medicines and therapies. The Government should be commended for this significant initiative that stands to provide relief for thousands of Canadian patients with rare diseases, many of whom do not have access to orphan drug therapies.

### "Rare" defined

Rare diseases are defined as those that affect fewer than 1 in 2,000 Canadians. They are frequently severe, life threatening, and progressively debilitating genetic diseases. 'Orphan' is the label applied to the drugs and therapies used in the treatment or relief of these rare disorders. And while the diseases are 'rare' in that each disease on its own impacts only a small portion of the general population, when combined these so-called rare diseases are anything but rare; they are numerous and affect thousands of Canadians and many thousands more globally.

Given the severe impact rare diseases can have on general populations, many G20 nations have already established orphan drug policy frameworks. These policies have an aim to nurture domestic rare disease research and development, and provide rare disease populations with increased access to available — and potentially new — therapeutic solutions.

The U.S. Congress passed the Orphan Drug Act of 1983 which provides support for the creation and development of medicines for rare diseases. Similar policy frameworks were introduced in Japan in 1993; Australia in 1998; and the EU in 2000. Canada needed to keep pace.

**Andrew Casey**
President and CEO, BIOTECanada

With its announcement Canada has begun the process of creating a national orphan drug policy framework that will align with frameworks found in other nations and will support 'made in Canada' therapies which will in turn provide relief for thousands of Canadians suffering from rare diseases.

### Establishing policies

For many years, Canadian biotechnology companies have been conducting significant research and development which has led to the development of new drug therapies that have fundamentally, and in some cases, permanently improved the lives of thousands of Canadian patients and many more globally that are afflicted with rare disorders. But more can and must be done as new rare diseases emerge each year.

By putting in place a competitive framework, the Government will establish a stable policy environment and provide the policy certainty necessary to attract investment. This investment is an essential component in driving increased orphan drug research and development in Canada. In this context, the industry is greatly encouraged by the initiative and looks forward to working with the Government to develop the final policy framework.

While this bodes well for the development of a strong Canadian biotechnology sector, ultimately it is those Canadians suffering from rare diseases that will benefit most as they will have greater access to a wider array of therapies and drugs.

### Biotechnology

BIOTECanada is the national industry association with nearly 250 members located nation wide, reflecting the diverse nature of Canada's health, industrial and agricultural biotechnology sectors. In addition to providing significant health benefits for Canadians, the biotechnology industry has quickly become an essential part of the transformation of many traditional cornerstones of the Canadian economy including manufacturing, automotive, energy, aerospace and forestry industries.

**ANDREW CASEY**
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# CHALLENGES

**FACT 1**
**WHEN COMBINED THERE OVER ARE 6,000 RARE DISEASES**

PHOTO: ACTUA

There are more than **6,000 rare diseases**, but parents have one feeling in common when they learn their child has a rare disease: **a feeling of being absolutely alone.**

# The challenge of rare diseases for Canadians

In most cases, parents are unaware of any history of rare disease in the family. Their child will have baffling symptoms that the doctor can't explain, repeated medical crises that the emergency room attendants dismiss and hundreds of tests with no conclusive results. Finally, when they receive the diagnosis, it's for a disease they can't even pronounce that has no cure and few treatments.

**Durhane Wong-Rieger, PhD**
President, Canadian Organization for Rare Disorders

### Rare diseases are really not all that rare

One in 12 of us lives with a rare disorder, which is roughly 2.8 million Canadians. That is about the same as the number of Canadians diagnosed with diabetes and more than twice as many as those living with all types of cancer combined. The challenge is that each rare disorder affects only a small number of patients, by definition, fewer than 1 in 2,000 persons, but in some cases, fewer than one in a million. About 80 percent are genetic, but some may be triggered by environmental factors. Additionally, 50 percent affect children and infants but for some disorders, symptoms occur later in life.

Prior to 1983, there were almost no treatments for rare disorders. In 1983, a patient coalition in the USA persuaded Congress to pass the Orphan Drug Act, providing incentives for developing drugs for "small and neglected" patient populations. Today, there are more than 400 rare disease drugs and over 2,000 in development. In 2003, another breakthrough was the sequencing of the 23,000 genes in the human genome, providing a base against which to identify genetic mutations causing rare disorders.

### Orphan drug policy

The Canadian Organization for Rare Disorders has led the advocacy for a Canadian orphan drug policy. On October 3rd, 2012, the Canadian government announced a regulatory framework to stimulate development and improve access to rare disease drugs.

And there has been other progress. In 2012, the Canadian Institutes for Health Research awarded the first research grants for rare disorders, $15.5 million for nine projects over five years. With the support of Genome Canada and others, two large research consortiums, IGNITE in Halifax and FORGE in Ottawa, are identifying genes that cause rare diseases and matching them with existing therapies. CORD is proud to be a sponsor and partner in many of these collaborations.

### Working together

Our next goal is the creation of a national plan for rare disorders that will include Centers of Reference for diagnosis and treatment, national guidelines for newborn screening to detect treatable and preventable rare diseases, professional education especially for family physicians and pediatricians, public awareness, and support for patients and families.

At CORD, we recognize that living with a rare disorder can be challenging for individuals and families but by working together to address the challenges facing all Canadians with rare disorders, we must work together. In 2011, CORD sponsored the Arctic Quest, a walk to the Arctic Circle not only to raise awareness of the capabilities of those with rare diseases but also to challenge all Canadians to join with us in our quest to improve the lives of all with rare disorders.

**DURHANE WONG-RIEGER, PHD**
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# How ERT helps patients with MPS

**Mucopolysaccharide (MPS) diseases are debilitating genetic disorders, collectively affecting approximately 1:25,000 Canadians — most of whom are children.**

**Kirsten Harkins,**
Executive Director, MPS Society

These affected individuals are born without particular enzymes necessary for normal cell degradation and recycling substances (mucopolysaccharides, now called glycosaminoglycans).

These substances are stored throughout their bodies, causing devastating progressive damage to their hearts, bones, joints, respiratory systems and, in some cases, brains and central nervous systems. While babies affected with MPS or a related lysosomal storage disorder often show no signs of the disease, symptoms appear and intensify as storage increases. As a result, many affected children never reach adulthood.

### Unlocking the mystery

In 2003, the first enzyme replacement therapy (ERT) for MPS (type I) was approved by the FDA, and in 2004, it was licensed for use in Canada. In 2005, an ERT for MPS II was approved, and in 2006, yet another ERT came on the market for MPS VI. Now, there's a clinical trial nearing completion for MPS IV A, and one on the horizon for MPS VII.

Although, there have been huge advances in research over the past decade and there are countless dedicated scientists working diligently to unlock the mysteries of these insidious disorders. MPS diseases are yet to be completely understood and there are several types of MPS with no effective treatment.

### Moving forward

While ERT halts most of the progression of the disease, symptoms remain and complications persist, even after missing enzymes are introduced. Early diagnosis is critical but challenging, and even once a diagnosis is made, accessing treatment can be a huge challenge. Often families have to fight for funding, even as they deal with the grief associated with the devastating diagnosis of a child, and the stress of living with a chronic disease.

Founded in 1984, The Canadian Society for Mucopolysaccharide and Related Diseases Inc. (Canadian MPS Society) is committed to providing support to affected individuals and their families. It educates medical professionals and the general public about MPS, and raises funds for research so that one day there will be cures for MPS diseases. For more information on MPS and the Canadian MPS Society, please visit www.mpssociety.ca, email info@mpssociety.ca, or call 1-800-667-1846.

**MEET JASPER**
**A little boy with MPS VI who is benefitting from weekly enzyme replacement therapy.**
PHOTO: SUSAN BENOIT

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WE RECOMMEND

**When did Cancer become a rare disease?**
PAGE 7

Patients with rarer cancers face a series of enormous uphill battles, from establishing an accurate diagnosis, to finding and accessing expertise, to obtaining treatment.



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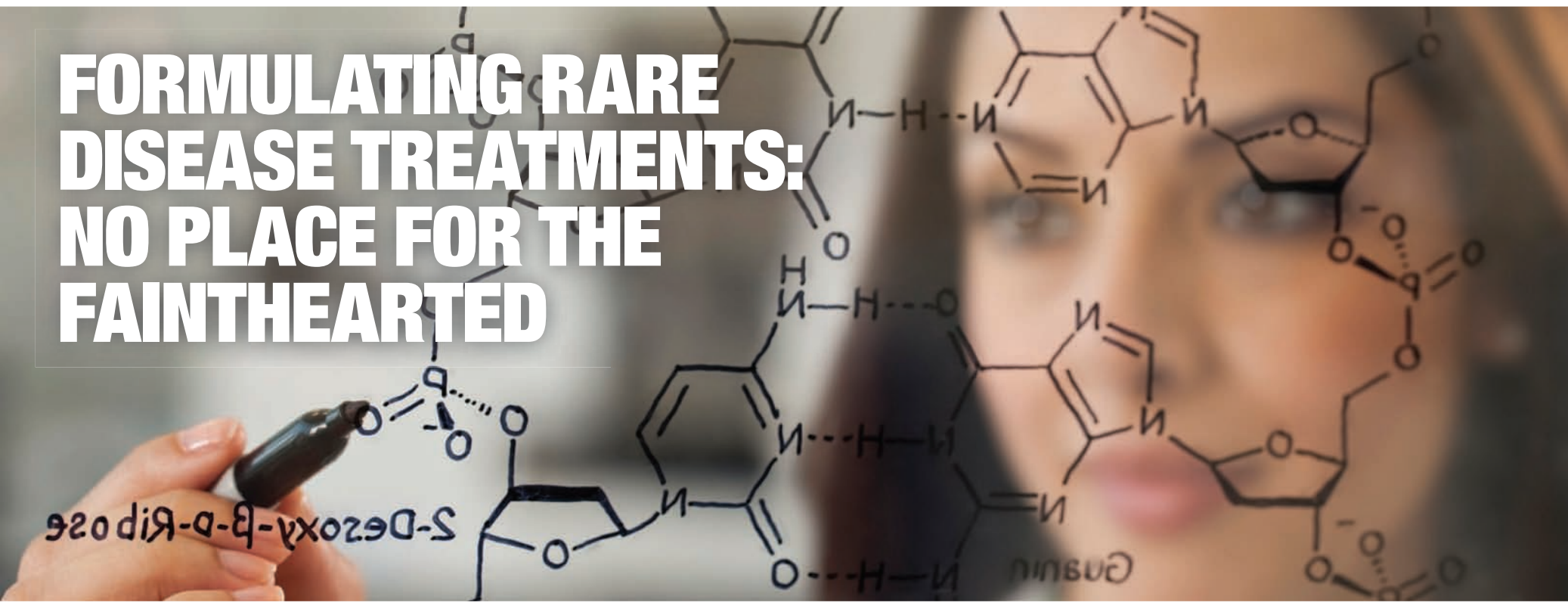
INSIGHT



# FORMULATING RARE DISEASE TREATMENTS: NO PLACE FOR THE FAINTHEARTED

## For Genzyme Canada General Manager Peter Brenders, surviving profitably in the rare disease space is impossible, **if your treatments are not effective.**

**"Our business' viability really comes down to the effectiveness of our treatments, and not the prevalence.**

We are not a company that goes after volume," says Brenders. "We focus on transformative therapies and our treatments do have a huge impact on people."

Founded in 1981, Genzyme has evolved from a tiny start-up to one of the world's largest biotech companies, with nearly 10,000 employees and operations worldwide. It was acquired by Sanofi in 2011.

Last year, Genzyme allocated $188 million (19 percent of total revenues) to research and development.

### Enzyme replacement therapies

The National Institutes of Health calculates there may be 5,000 to 6,000 rare diseases, about 4,000 of which are genetically based (single-gene mutation).

Genzyme's therapies are in the class of rare genetic diseases known as lysosomal storage disorders (LSDs). This is a group of about 50 rare metabolic disorders caused by deficiencies in a single enzyme required to metabolize substances like fats and sugar containing proteins.

"We have capitalized on enzyme replacement therapies. The company has gone through several products that have ebbed and flowed, but we have decided to stay true to our focus of being very specialized and not try to be all things to all people," says Brenders.

Today, Genzyme's core products are very small and very niche. It is focused on five disease areas — Gaucher, Fabry, Mucopolysaccharidoses (MPS), Pompe disease, and thyroid cancer management.

The prevalence rates for these diseases vary between the 1 in 40,000 to 1 in 100,000. "In LSDs, we have some 138 patients across Canada," Brenders offers.

"Every company says they are patient focused but we are a company that knows and cares for all our patients. It is companies like ours that have truly taken on the risk to further the knowledge in this space. This is what makes our work so exciting — to help where no one else wanted to go."

**Peter Brenders**
General Manager, Genzyme Canada

"Our business' viability really comes down to the effectiveness of our treatments, and not the prevalence."

Recently, Genzyme has expanded its focus to other rare diseases and multiple sclerosis. In September, the US FDA approved Aubagio (teriflunomide) as a new once-daily, oral treatment indicated for patients with relapsing forms of multiple sclerosis.

### Challenges remain

The Canadian Organization of Rare Diseases (CORD) says that 1 in 12 Canadians has a rare disorder. Many others are affected or at risk but remain undiagnosed and unaware.

Unlike many industrialized countries, Canada has inconsistent access to therapies for rare diseases. The question of whether a patient can access treatment depends on the province in which they live.

Orphan drugs, which have been developed for the treatment of rare diseases, which typically afflict a handful of patients, can be high cost, between $100,000 to $800,000 a year. Obtaining health insurance for treatments with so few patients can be remarkably difficult.

"There is no real portability of rare disease treatment. Because of this variance, patients are stuck, or even worse, neglected to suffer," Brenders notes. CORD has called on governments to address this with a national "Chance for Life" fund so that those with rare disorders can be equitably treated in Canada's health care system.

But Brenders is optimistic. "We are proud of our commitment to providing hope where there was none before. We remain hopeful that Canadians in need will soon have access to care."

**INDRANI NADARAJAH**
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# Diagnosing and living with Pompe disease

About 1 in 40,000 people suffer from Pompe disease, an inherited and often fatal disorder that severely weakens the heart and skeletal muscles. It is caused by mutations in a gene that makes an enzyme called acid alpha-glucosidase (GAA). GAA breaks down glycogen within lysosomes, a stored form of sugar used for energy.

Pompe disease is about five times more common in adults than infants. In adults, the symptoms first appear usually when patients are in their 20s. It takes another 10 years before it is diagnosed, because the disease progresses so slowly, says Dr. Mark Tarnopolsky, professor of paediatrics and medicine at McMaster University. Primary symptoms are usually muscular weakness which progresses to respiratory weakness.

Adult Pompe disease is usually challenging to diagnose. The fairly mild symptoms in the early stages are sometimes confused with fibromyalgia or chronic fatigue syndrome. Pompe Patients can be misdiagnosed with more common rheumatologic or neurologic conditions such as polymyositis or muscular dystrophy.

**PATIENT AND DOCTOR**
**LEFT: Dr. Mark Tarnopolsky, Professor of Paediatrics & Medicine, McMaster University**
**RIGHT: Velda Lachance, diagnosed with Pompe disease**

"People often frequently self-diagnose, blaming age or overwork for their tiredness, so this leads to further delay in diagnosis," Dr.Tarnopolsky notes.

"I diagnosed my first [Pompe] patient about 16 years ago, and there was nothing much to offer them then." Today, though, patients are treated with an enzyme replacement therapy called Myozyme. Myozyme is extremely effective in children, reducing fatality rates from 75 percent to less than 10 percent in the first year, and from 90 percent to about 20 percent in the second year of life. While improvement in older patients is not so dramatic, the rate of muscle strength loss and breathing difficulty is slowed, Dr.Tarnopolsky explains.

Many doctors have become more aggressive about identifying and treating nutritional deficiencies, especially vitamin D and B12, in patients with neuromuscular disorders such as Pompe disease. Recent data also confirms the importance of exercise in managing this disease.

"We have found that careful management of exercise and/or physiotherapy and optimizing nutrition are very important in helping to manage a wide variety of genetic disorders."

### The patient's perspective

Velda Lachance, 52, first noticed her symptoms in her late teens, when going up and down stairs became a struggle.

Unfortunately, it was downhill from there.

At 25, she was diagnosed with fibromyalgia, the first in a long list of misdiagnosis, which also included arthritis and osteoporosis. She became very sensitive to touch, and couldn't sleep on her left side, because "it took her breath away".

Lachance deteriorated from a person who was extremely active and sports-mad in school, to one who lived in a wheelchair. Because her diaphragm was paralyzed, she couldn't cough or sneeze. "Some of the doctors implied that I was imagining things and I was told it was all in my head," she says matter-of-factly.

She had practically given up hope of finding any answers when she was diagnosed by Dr. Tarnopolsky three years ago.

### Life can get better

The right diagnosis has given her her life back. Today, Lachance has a Myozyme infusion every two weeks, which she credits for helping her feel much better. "I call it my lifeline," she jokes.

She's stopped smoking. She exercises religiously at the gym, and is eating more healthily. "I work out with arm and ankle weights, and I am strengthening my core which has become very weak from being in a wheelchair. My breathing has improved.

"My goal is to walk again," she confides. She also meets her friends for coffee, a far cry from when she was housebound.

Her diaphragm is still paralyzed and she suffers from sleep apnoea, but Lachance doesn't dwell on those problems.

"I was in a dark place for 27 years but life is so much better now. To those who have been told they're imagining things — don't ever give up."

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INSPIRATION

FACT 2

THERE IS NO STANDARD CANADIAN DEFINITION FOR RARE DISEASES

Rare genetic disorders affect about **250,000 children** in this country, but improved technology and funding are helping Canadian researchers to **unlock some of the body's darkest secrets.**

# THE QUEST TO IDENTIFY RARE DISEASES

## SEARCHING FOR ANSWERS

There are about 7,000 "orphan", or rare diseases, but little is known about them, despite its huge impact on families, say doctors.

"When we walk around the paediatric wards, roughly a third of the children have orphan diseases. There's a huge unmet medical need," says Dr. Alex MacKenzie, Professor of Paediatrics at the University of Ottawa.

There is no standard Canadian definition for rare disease, but these 7,000 diseases have a prevalence rate of 1 in 50,000 or less.

Dr. Kym Boycott, a medical geneticist and researcher from the Children's Hospital of Eastern Ontario, says it is very difficult to tell parents of a child with complex medical issues that "we know your child has a rare genetic disease, we just don't know the name of it.

"These families often embark on a 'Diagnostic Odyssey' punctuated with MRIs and biopsies, for the next four to five years to try to discover answers," she says. "Unfortunately, there is no guarantee that we will arrive at a diagnosis at the end of it."

### Next-generation sequencing

However, a new type of DNA sequencing, termed next-generation sequencing, has revolutionized the study of rare genetic diseases, allowing a person's entire genetic code of about 22,000 genes to be rapidly analyzed within a few days. It is now possible to find disease-causing genes using a relatively small number of patients more cost-effectively, explains Dr. Boycott.

Then, in April 2011, the Canadian scientific community was bestowed $4 million of public research dollars — from a collaborative group including Genome Canada, Canadian Institutes of Health Research, Genome Quebec, Genome British Columbia, the Ontario Genomics Institute and the McLaughlin Centre — to investigate the origins of rare diseases.

The excitement, as Dr. MacKenzie describes, was immense. "Within months of the money being awarded, there was a linking of hands across the country to try to find answers to some of the more perplexing diseases."

**Dr. Alex Mckenzie**
Professor, Paediatrics
University of Ottawa

"When we walk around the paediatric wards, roughly a third of the children have orphan diseases. There's a huge unmet medical need."

### FORGE Canada

FORGE Canada (Finding of Rare Disease Genes), launched on April 1, 2011, is a national consortium of clinicians and scientists using next-generation sequencing technology to identify genes responsible for a wide spectrum of rare pediatric-onset disorders in the Canadian population. It brings together clinicians from all 21 clinical genetics centres in Canada, and clinicians from 17 countries.

**Dr. Kym Boycott**
Medical Geneticist & Researcher
Children's Hospital of Eastern Ontario

"Families often embark on a 'Diagnostic Odyssey' punctuated with MRIs and biopsies, for the next four to five years to try to discover answers."

Requests for proposals have yielded over 200 disorders that meet FORGE criteria. These disorders range from those affecting single families to disorders with over 20 patients from across Canada and internationally.

FORGE has investigated 132 diseases and identified 77 genes, of which 37 are brand new disease genes.

### The quest for therapies

Naming a rare disorder is important, but the Holy Grail lies in developing cost-effective therapies that work.

Currently, 200 of the 7,000 diseases have treatments, some of which are effective, others not so. "It's a bit of a mixed bag," says Dr. Boycott, who heads up the FORGE team. The current funding runs out next April, but FORGE has already applied for a $12 million grant, which will be directed towards developing more therapeutic approaches, she says.

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# Education and early detection is crucial for those with Fabry disease

## ABOUT FABRY DISEASE

Fabry Disease is an inherited metabolic disorder where the body does not produce sufficient quantities of an enzyme called alpha galactosidaseA ("a-gal A").

This enzyme is used to break down a naturally occurring substance that the body produces called globotriaosylcera-mide ("Gb3"). This substance accumulates throughout the body, damaging the internal organs, the heart and brain, and shortens lifespan.

There are approximately 350 known cases of Fabry Disease in Canada (about 1:100,000). Since the Gb3 accumulates in cells throughout the body, it impacts all bodily functions.

The majority of patients suffer mild to extreme gastrointestinal problems, pain, heat intolerance, loss of function kidney, heart disease, and strokes. Without treatment, the average patient experiences kidney failure in their early 40's and a life span of around 50 years.

### Patients face many challenges

Fortunately a treatment option for this rare disorder was introduced in the early 2000's called enzyme replacement therapy ("ERT"). With ERT, a manufactured enzyme is intravenously infused over about two hours into the blood stream every two weeks. If treatment begins before organ damage occurs, then patients can hope for a normal life span.

Fabry Disease patients face many challenges. Many patients suffer years of misdiagnosis due to the rarity of the disease and symptoms that involve all systems of the body. The gastrointestinal problems and pain can lead to depression and unemployment challenges. This in turn can lead to social isolation.

With treatment, patients can lead normal, productive lives.

### Support

The Canadian Fabry Association ("CFA") is a volunteer association formed in 2005 to assist Canadian's with Fabry Disease by providing education, access to treatment and a social network.

DARREN BIDULKA,
PRESIDENT,
CANADIAN FABRY ASSOCIATION
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REACHING NEW HEIGHTS

INSIGHT

## Anything is possible

On August 2011, one dozen Canadians with rare disorders set forth on an extraordinary quest. Their destination: the Arctic Circle.

Their goal: to show that people with rare disorders can do just about anything...with the right treatment and support.

SEBASTIEN LAROSE
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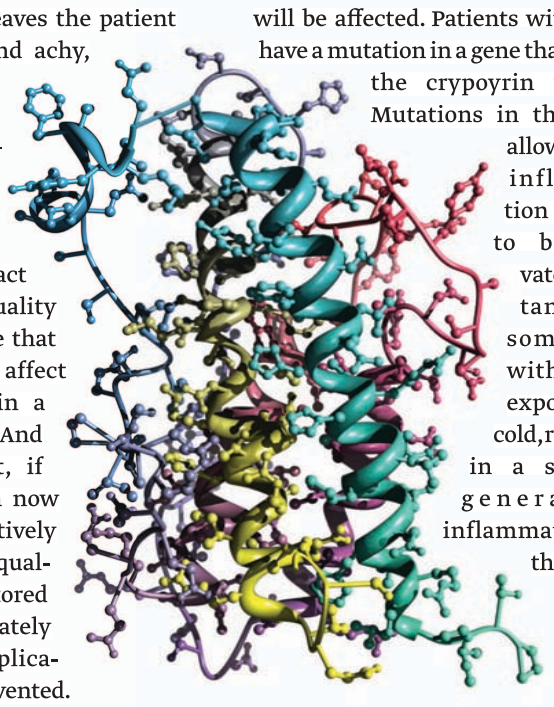
NEWS

# An extrodinarily unfamiliar rare disease

**Imagine having a disease that almost no physician has heard of.**

A disease that affects many tissues of the body and leaves the patient feeling tired and achy, virtually all the time. A disease with potentially life-threatening complications and severe impact on patient's quality of life. A disease that is reported to affect only about 1 in a million people. And a disease that, if recognized, can now be treated effectively such that the quality of life is restored almost immediately and the complications can be prevented.



Such is the life for patients with CAPS, the Cryopyrin Associated Periodic Syndrome. Cryopyrin is the name of a protein that is activated by cold temperature or slight reduction in temperature. Periodic Syndrome implies that the patient's symptoms are not necessarily present all the time, but may only be present in a periodic way.

CAPS is inherited in an autosomal dominated fashion, meaning that 50 percent of patients with the disease will be affected. Patients with CAPS have a mutation in a gene that makes the crypoyrin protein. Mutations in this gene allow for the inflammation system to be activated spontaneously, sometimes with mild exposure to cold, resulting in a state of generalized inflammation of the body.

## Cryopyrin Associated Periodic Syndrome

CAPS is not just one disease, but rather three overlapping diseases on a spectrum that ranges from mild to severe.

■ The mildest end of the spectrum, FCAS (Familial Cold Autoinflammatory Syndrome) is marked by very early onset (almost all within the first six months) of a hive-like rash, low grade fever, fatigue, aches and often red eyes. Patients report almost daily symptoms which are often brought on by slight change in temperature. In fact, they may be so sensitive to slight reductions in temperature that they avoid movie theaters and shopping malls for fear of precipitating an attack. FCAS does not reduce longevity but does have a significant impact on patient's quality of life and vitality.

■ At the most severe end of the spectrum is a disease called NOMID (Neonatal onset multisystem inflammatory disease). As the name suggests, this disease has its onset in the newborn (neonatal) period. There is a very early onset fever, hive-like rash, severe irritability, often difficulty with feeding, and inflammation of the lining of the brain (meningitis), inflammation of the hearing mechanism which may lead to deafness, and inflammation of the nerves that supply vision which may lead to blindness. Inflammation may also occur around some of the large joints resulting in significant deformities. Many children previously affected with NOMID have severe delayed development, deafness, blindness, and severe arthritis.

■ The disease Muckle-Wells Syndrome (MWS) is the middle of these two in terms of severity. Short attacks of fever, hive-like rash, joint pain, and red eyes usually start at childhood. Approximately 50 percent of these patients develop severe hearing loss.

As a result of the ongoing inflammation in both Muckle-Wells Syndrome and NOMID, many of the proteins that are made during inflammation may deposit in specific organs resulting in a long-term complication called amyloidosis. This usually presents changes in the kidney function which can ultimately result in kidney failure needing dialysis and even transplantation.

## Testing and diagnosis

Up until the last few years, there were no treatments that could consistently reduce and prevent the spontaneous attacks of inflammation suffered by patients. Over the last decade drugs of the biologic class have been developed that counteract the effects of interleukin 1, the key inflammation protein that is elevated in CAPS.

Research has shown that these drugs can not only prevent attacks, but may also reverse some of the more serious disease manifestations which were previously thought to be irreversible. But patients will only be able to receive these new drugs if someone recognizes that they may have CAPS and order the appropriate testing.

**Ronald Laxer**
Professor, Paediatrics & Medicine,
Univeristy of Toronto;
Staff Rheumatologist,
The Hospital for Sick Children

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NEWS

# WHAT WE DON'T KNOW CAN HURT US WHEN IT COMES TO FIGHTING ILLNESS

**Survey shows that better awareness and education is needed to improve care for Canadians living with rare lung disease**

**Robert Davidson**
President,
Canadian Pulmonary Fibrosis Foundation

Unlike Canadians living with common diseases, people living with little-known diseases face challenges associated with a lack of knowledge among patients, their caregivers and health care providers. Unfortunately for these people, such challenges include delayed diagnosis, limited access to specialized care, and poor availability of safe and effective treatment options.

This is certainly true in the case of idiopathic pulmonary fibrosis (IPF), a rare, progressive and life-limiting lung disease with no known cause. According to a recent survey of IPF patients in Canada, "An Investigation into Patient Experiences with IPF", what you — and your health care provider — don't know can hurt you.

As indicated in the survey, greater awareness is needed to hasten diagnosis of the disease. Patients wait almost 20 months from when they first begin to experience symptoms to receive a confirmed diagnosis of IPF, and only 32 percent of patients receive IPF as their first diagnosis.

## Access to treatment

Given the disease is rapidly fatal, even one day waiting to see a doctor in order to be diagnosed and referred to the proper specialist, or to get access to effective treatment, is one day too many.

Patients without access to experts in specialized care clinics across the country told us they are dissatisfied with the current level of care they receive. Further, the lack of access to new, safe and effective treatment options for IPF is a great cause of concern for patients, given the high dissatisfaction with currently available therapies.

## Fighting for change

The Canadian IPF community has spoken through this patient survey — we don't accept the current paradigm, and we know what needs to be done to give us a fighting chance at improved quality of life and life itself.

For anyone who has been diagnosed with a disease — rare or otherwise, it is important to get informed so you can take control of your own health and win the fight. In the words of Winston Churchill, "Never surrender."

ROBERT DAVIDSON
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# Home health care

**The complexity of many rare diseases dictates that sufferers require specialized health care in their homes.**

Whilst this type of care can be expensive, it can have wide ranging positive effects for the patient and, in a broader sense, the medical industry.

Zoe Vernham, a manager in business development in the home health care industry, has an abundance of experience in community nursing working in some of Canada's most challenging environments.

## Increased comfort and care

She explains: "Giving home health care really helps out the hospitals as it deals with a lot of the overflow of patients who, instead of having to go into hospital, get their care at home. It means that demand in hospital is eased up and that patients in hospital get a higher level of care."

Vernham believes that the level of care received by patients who get home health care can increase their quality of life immeasurably: "Patients are most comfortable in their own home. Being surrounded by family and being in familiar surroundings with their own belongings is great for wellness and recovery."

She feels that home care can really lift a patient's self-esteem: "They are able to care for their appearance and wear their own clothes and jewelry. They feel a real sense of dignity. This is something that is so important for the wellbeing of patients."

## Ensuring proper treatment

A large aspect of home health care is teaching the patient and their family how to administer medicines. This education is vital because, in many cases, drugs need to be injected by the patient themselves and this can difficult for the patient to get adjustto.

Vernham is confident that having a health care professional come into the home makes a significant difference: "Statistically, patients learn a lot better if they are in their home environment with a care partner or a close family member."

Vernham's enthusiasm for the benefits of home health care is unmistakable: "There are so many positive challenges, it is such a rewarding job. Not only do you make a big difference in the patients' lives, they make a big difference in yours."

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INSIGHT

FACT 3

CANCERS CAN BE CONSIDERED A RARE DISEASE

LEADING A NORMAL LIFE Dave Galley and his wife Ingrid enjoy a walk in the park.

# Dave Galley details living with a rare blood cancer

After surviving a brain tumor (an Acoustic Neuroma) and getting back on to his feet after 12 hours of brain surgery that successfully removed most of it Dave Galley, who just turned 50 thought that his health issues were now over and he could get his life back on track. Apart from the odd MRI to check that no regrowth is occurring, which it has not, Dave resurrected an overdue knee surgery to get his sporting life going again.

So it is with a hint of irony that the blood test that is a standard part of the pre-op procedure revealed an anomaly, which was first brought to Dave's attention by a telephone message left on his cell to call the office of Dr Tailor; a haematologist. Dave returned home from that appointment to inform his family he has just been diagnosed with Leukemia.

David explained to his family that he has CML (Chronic Myelogenous Leukemia) a rare blood cancer.

### Understanding the diagnosis

Having come to grips with the diagnosis and the fact that there were very good treatment options for CML Dave and his family (consisting of his wife Ingrid and 2 teenage sons Charlie and Jack) got on with their lives until Dave attended Dr. Tailors clinic for a bone marrow aspiration to confirm CML.

Dr. Tailor changed his diagnosis from CML to MF Myelofibrosis, a form of Leukemia, which at the time had no reliable treatment.

The best hope was Stem Cell Replacement, a high risk and donor-dependent treatment. Although, within minutes of the diagnosis, Dave was then informed of a clinical trial for MF that was starting that very same week. The trial was being lead by Dr. Van Der Jaqt, a haematologist at the Ottawa Civic hospital. Further analysis showed that Dave's cancer was triggered by the JAK 2 inhibitor, a factor that the clinical trial was targeting.

As the seriousness of MF settled into Dave and his family's minds, especially when traversing through the process of applying to go on the clinical trial, they went through what they describe as the lowest period of their lives.

Knowing the trial offered hope, and knowing what was coming down the road with the eventual debilitating final stages of the cancer; but not knowing if Dave would be on the trial was just about as tortuous as it gets as far as they were concerned. It was all the family could focus on whilst they tried to deal with the situation.

Shortly after this period the most wonderful period for the very family followed. Within weeks of starting the clinical trial for the drug, Dave's symptoms started to disappear.

The symptoms that Dave now knows with a clarity that is only available with hindsight were indicative of a serious illness. These were night sweats, weight loss, itchy red rashes, aches and pains, mild flu like symptoms, stomach cramps due to an enlarged spleen and a constant lethargy that was all put down to stress and age.

### Recovery

Now, over 2 years later and still receiving the drug, Dave is leading a normal life. There are no side effects and Dave's blood tests are all in the normal range. Dave is aware that the drug does not cure MF; the drug hunts down the damaged cells and turns them off, therefore keeping Dave in remission. Dave hopes this will be as effective as the drug used by CML sufferers, who after 12 years on are still leaving normal lives and still taking their drug.

He hopes that it is rapidly approved and accepted across the worlds health authorities and medical health companies such that it is readily available to all who are unfortunate to suffer from this extremely rare and debilitating disease.

**DAVE GALLEY**

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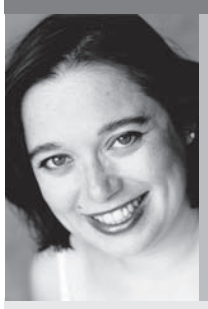
CHECK THIS OUT



**Isabel Jordan**
Chair,
Rare Disease
Foundation

### "Parent 2 Parent"

People gather around a large table, some greeting each other like old friends, others new to the group. When a speaker addresses the gathering and asks them to introduce themselves, it seems their reasons for attending are widely varied. Yes, they are all parents, but each of their children suffers from a profoundly different medical condition. Each child faces different physical and mental challenges on a daily basis — not one story is the same.

Yet as the meeting progresses, the commonality within the group becomes obviously apparent. The challenges vary, but the life experience of dealing with a rare disease diagnosis is something they all share. And together they deal with the ever-present uncertainty that rare disease brings by drawing on each other for strength, advice, resources, and even laughter.

### A support network

So goes a meeting of the Rare Disease Foundation Parent 2 Parent Resource Network. Initiated four years ago in Vancouver as an experiment in peer-to-peer support, the goal was to develop a support network for those that had none. As a result, for the first time many participants could voice their fears, address their uncertainties, and help each other navigate the complex pathways of the medical, academic, and social systems - all in search of better solutions for their children.

With the success of the initial model, and with 1 in 12 Canadians being affected by rare disease, outreach to new parents has continued. Today the Rare Disease Foundation supports Resource Network meetings in Vancouver & Ottawa, as well an online forum for all parents. New groups are currently launching in Toronto & Winnipeg as well as being planned for smaller locales. The vision is for all parents to feel they don't have to walk these uncertain paths on their own.

For more information on existing and new Parent 2 Parent Resource Network groups contact families@rarediseasefoundation.org.

**ISABEL JORDAN**

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DID YOU KNOW?

NEWS

## An introduction to Gaucher Disease

Gaucher disease is a rare inherited genetic condition. Individuals with Gaucher disease are lacking or have insufficient amount of an enzyme that is needed in the body to breakdown a fat molecule called glucocerebroside. The body accumulates this fatty substance causing a build up in the spleen, liver, bone marrow, and other organs. This accumulation can cause symptoms such as bone pain, anaemia, fatigue, bruising, bleeding, and enlarged spleen. The bone symptoms can result in severe pain, damage to joints, and fractures.

**There are currently three recognized types of Gaucher Disease.**

■ Type 1 is the most common and does not affect the brain or nervous system. Some patients with Type 1 Gaucher disease have no symptoms, while others develop serious symptoms that can be life threatening.

■ Type 2 is very rare and usually has severe neurological symptoms. Children develop signs and symptoms within the first year of life. Many do not live past age two.

■ Type 3 may also cause neurological signs and symptoms, but they are less severe than in Type 2 Gaucher disease. Signs and symptoms appear in early to late childhood, and patients with Type 3 Gaucher disease live well into adulthood.

The National Gaucher Foundation of Canada is a voluntary organization that supports families and individuals afflicted with Gaucher Disease. Our primary mission is to improve the health and wellbeing of Canadian Gaucher Patients.

If you have any questions or would like more information on Gaucher Disease, please visit our website at www.gaucher-canada.ca.

**CHRISTINE WHITE**
**PRESIDENT,**
**THE NATIONAL**
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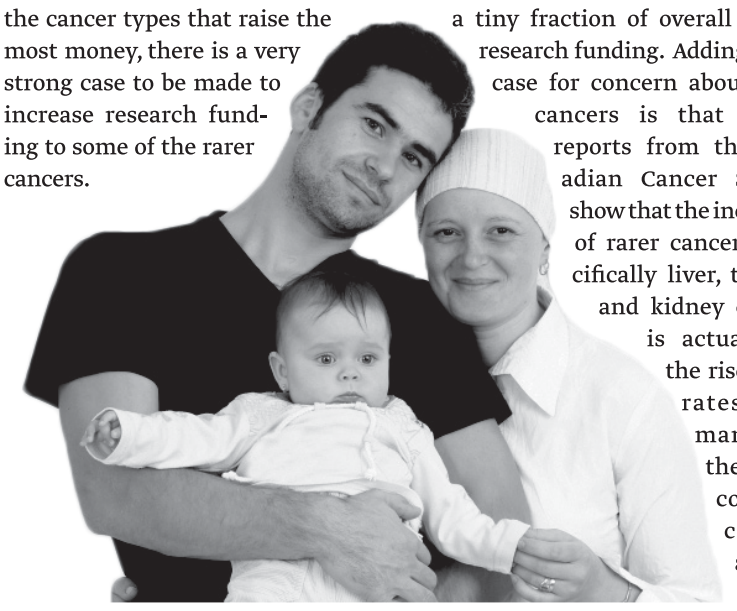
# When did "cancer" become a rare disease?

**With so many high-profile walks, runs, and mega-fundraising events for some of the more recognizable cancers, some might conclude that "cancer" research and cancer treatments are well-funded in Canada. This conclusion, while incorrect in the broadest sense, strikes painfully far from reality for those of us with rarer cancers.**

While it is no surprise that research funding in Canada focuses most on the cancer types that raise the most money, there is a very strong case to be made to increase research funding to some of the rarer cancers.



While lung, prostate, breast and colorectal cancer, the big four in the cancer world, account for over 50 percent of all new cancer cases in Canada, there are dozens upon dozens of other cancer types that make up the remaining 50 percent. Over half of all deaths from cancer in Canada are attributed to cancers other than the top four. Many are diagnosed at a late stage and have few, if any, funded treatment options.

Despite their higher death rates, rarer cancers such as pancreatic cancer and kidney cancer receive only a tiny fraction of overall cancer research funding. Adding to the case for concern about rarer cancers is that recent reports from the Canadian Cancer Society show that the incidence of rarer cancers (specifically liver, thyroid, and kidney cancer) is actually on the rise while rates of many of the more common cancers are falling.

### An uphill battle

Patients with rarer cancers face a series of enormous uphill battles, from establishing an accurate diagnosis, to finding and accessing expertise, to obtaining treatment. Disease-specific patient organizations such as Kidney Cancer Canada and others will continue to advocate and raise funds for more research. More than just fundraisers, these small patient-focused organizations provide patients with a lifeline for information and vital help to navigate within a complex cancer system.

In the emerging era of personalized medicine, we hope to see all cancers treated for what they are: a genetic mutation versus a disease that happens to arise in a specific organ or body part. We look forward to a day when we will have better answers for everyone diagnosed with cancer in their lifetime.

In the meantime, if we are to save unnecessary deaths from cancer, we need to focus our attention on those cancers that most urgently need our time, attention, and research investment. A rarer cancer presents a rare opportunity to make a significant difference.

**DEB MASKENS**

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