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PRESIDENT'S REPORT



CORD President,
Durhane Wong-Rieger

As president of the Canadian Organization for Rare Disorders, I am truly optimistic that 2007 will be heralded as the year Canada began to fulfill its obligation to patients with rare diseases and disorders. In 2007, we brought together patients, clinicians, researchers, industry, and government in regional forums and a national/international conference to discuss the proposal for a Canadian orphan products program that would provide access to innovative therapies for patients with rare conditions as well as support the development of new therapies in Canada.

CORD proposed the Chance for Life Fund, which would make Canada the international leader in rare disorders. By allocating an amount equivalent to 2% of the public drug funds, we could assure that patients with severe, debilitating and life-threatening disorders will have a fair chance to treatments that enhance quality of life and, in many cases, provide the only chance for survival.

Someone asked why treatments for rare diseases/disorders were known as orphan drugs. The answer is, "Because no one wanted them." Potential treatments were considered too difficult and expensive to develop because of the very small patient population. It was too difficult to conduct clinical trials, too difficult to demonstrate outcomes, and too difficult to generate any return on very high development costs. In the decade prior to 1983, there were only 34 new drugs for rare disorders. In the 25 years following the US Orphan Drug Act, there have been more than 300 new drugs brought to market for rare disorders and over 1700 new entities registered as potential therapies. In 1993, Japan enacted orphan drug legislation and in 2000, the European Union passed Orphan Drug legislation for all 27 member states. Worldwide, rare disorders are becoming orphans no longer.

At the 2007 European Organization for Rare Disorders Conference, a ministry representative said she had been asked, "Why should we invest in diseases that affect so few people?" She replied, "Beyond the obvious reason that persons with rare disorders deserve the same right to healthcare as anyone else, we invest because we are learning so much with rare disorders." Today, orphan drugs involves the most innovative and exhilarating research, which will have benefits far beyond rare disorders. Moreover, as genetically targeted therapies such as those available for various forms of cancer become more prevalent, all therapies will eventually become "orphan" drugs.

CORD hopes that Canada will soon shed its ignoble distinction as the "only developed country without an orphan drug policy." In 2005, the Federal/Provincial/Territorial Ministers of Health made a commitment to fund "Expensive Drugs for Rare Disorders." We were pleased that the Ministers finally recognized the need for a unique program for the "rarities," but we have been discouraged by the very limited scope of the proposed program and the slow progress. One meeting in two years is not adequate when patients are literally dying for lack of access to therapies that are already available in many other countries.

CORD salutes the initiative of Don Bell, MP from North Vancouver, who recently introduced a motion in the House of Commons to adopt an official definition of "rare disease", defined in most countries as conditions with a prevalence of less than 1 in 2,000. We are optimistic following our meetings with federal Minister Tony Clement and his staff, as well as meetings with various provincial ministry representatives. We are grateful for the active involvement of the CORD Corporate Council and our many doctors, nurses, and other health professionals. We urge all Canadians to call upon their political representatives to give the Chance for Life to all patients with rare diseases and disorders.

On February 29, 2008, CORD in collaboration with rare disorders groups across Canada will host the 1st International Rare Disease Day activities. Please join us for a planned rally in Ottawa on February 28, as well as activities in provincial capitals and other locations across the country.

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Treatment of rare disorders: U.S and Europe enact legislation to encourage innovation and access while Canada lags behind.

The European Commission, European Medicines Agency and the U.S. Food and Drug Administration recently adopted a common application form for drug makers seeking orphan designation for their medicines. This will simplify the drug approval process and spur innovation by allowing companies to apply for approval of their new product in both the US and across Europe at the same time. In addition, this new process will help the regulatory agencies to

better understand each other's systems. [1]

In contrast, patients and advocates in Canada have expressed concern that the Common Drug Review (CDR) – a government appointed agency that makes

recommendations to provinces (with the exception of Quebec) regarding listing decisions for new drugs – is failing to provide patients with timely access to new medicines and failing to operate in an accountable fashion. The CDR process has recommended against reimbursement for every treatment evaluated for unmet needs.

In response to these concerns, the federal House Standing Committee on Health recently conducted a review of the CDR. The government recommendations include: [3]

- Establishing a specifically designed approach for the review of drugs for rare disorders and for first-in-class drugs;
- Creating a distinct appeal process with a separate group of experts;
- Increasing the current level of public involvement in the CDR through public attendance at open CEDAC meetings and the creation of a public advisory body.

International Comparison of Public Reimbursement of Products for Unmet Needs [2]						
BRAID ^a	INDICATION	Canada (CDR)	UK	France	Sweden	USA (Medicaid)
Somavert	Acromegaly	No	Yes	Yes	Yes	Yes
Xolair	Severe Persistent Asthma	No	Yes	Yes	Yes	Yes
Aldurazyme	Enzyme Replacement Therapy	No	Yes	Yes	Yes	Yes
Fabrazyme	Fabry Disease	No	Yes	Yes	Yes	Yes
Sensipar	Chronic Kidney Disease	No	Yes	Yes	Yes	Yes
Forteo	Osteoporosis	No	Yes	Yes	Limited	Yes
Replagal	Fabry Disease	No	Yes	Yes	Yes	Yes

^a Products Licensed in Canada, US, and EU

While these recommendations are encouraging, this review is non-binding and merely suggests that the CDR consider these suggestions. The extent to which CDR implements these, and other Committee recommendations, remains to be seen.

Rare Disorders

Despite the small numbers affected by each disease, there are up to

6,000 rare disorders which affect nearly 10% of the Canadian population, 30 million people in Europe and about 25 million Americans.

[1] http://today.reuters.com/news/articleNews.aspx?type=scienceNews&storyID=2007-11-26T183432Z_01_L2677301_RTRUKOC_0_US-DRUGS-REGULATORS.xml

[2] BIOTEC Canada – Presentation to the House Standing Committee on Health (April 16, 2007)

[3] House Standing Committee on Health Report on the Common Drug Review (Dec. 2007) <http://cmte.parl.gc.ca/cmte/CommitteePublication.aspx?COM=13189&Lang=1&SourceId=220278>

Toward a Canadian Chance for Life Fund

Report of 2006-07 Forums and Conferences

For Canadians with rare diseases and disorders, 2007 was a year of unprecedented activity...and hope. The year began with the second in CORD's series of forums toward a Canadian Orphan Products Policy.

Ontario OPP Forum I (Access)

The inaugural forum in the OPP series was held on October 24, 2006 in Toronto and was directed to raising awareness of the inadequacy of the current drug funding review process for rare disorders and to generate recommendations for improving access. We were fortunate to have in attendance Helen Stevenson, Executive Lead for the Ontario Drug Strategy Secretariat and now Assistant Deputy Minister and Executive Officer of the Ontario Drug Program. Among the many highlights from this forum were the address from Andreas Laupacis, former Chair of the Canadian Expert Drug Advisory Committee (for the Common Drug Review) and the provocative discussion led by Professor Amir Attaran from the Institute of Population Health at the University of Ottawa on Ethical and Legal Issues related to access to treatments for patients with rare disorders. Dr. Laupacis acknowledged that the Common Drug Review was not the appropriate body for reviewing drugs for rare disorders. He stated, categorically, that the CDR did not even want to receive the file for Fabrazyme (for the treatment of Fabry's Diseases) because it was clear that the drug would not meet standards of cost-effectiveness. But the provinces insisted, so they reviewed it, with predictable rejection.

Professor Attaran set for the opinion that the current (closed meeting) process of drug review was in violation of the Canadian Charter of Rights and Freedoms, which guaranteed no person to be arbitrarily denied life, liberty or security of person. Among the key issues identified by participants, who included representatives from patient organizations, medical professions, and industry, were the recognition that (1) Canada lagged the rest of the developed world in treating rare disorders patients; (2) the Canadian process based on pharmacoeconomics inevitably failed patients with rare disorders because it was based strictly on cost and did not take into consideration societal values or ethics; (3) an "Expensive Drugs for Rare Disorders" program should not be based on the Fabry's and MPS I "research study" funding model; and (4) patients must be directly involved

in all aspects of developing and negotiating programs for the treatment of rare disorders.

Vancouver OPP Forum 2 (Innovation)

The key objective of the Vancouver forum, held February 12, 2007, was to explore options for improving Canada's capacity to develop innovative technologies for rare disorders and orphan conditions. The forum directly challenged Health Canada's position that there was no need for a Canadian orphan drug policy because Canadian patients with rare disorders already had access to new drugs. Drugs were already being developed in other countries; Canadian companies could take advantage of currently available business development incentives; and Health Canada was able to expedite review for life-saving drugs for rare disorders under its existing process. The multi-stakeholder forum supported the white paper from BioteCanada that called for Canada to join the rest of the developed world in providing market exclusivity, fee waivers, grants, tax credits, protocol assistance, and expedited reviews to stimulate development of new orphan products.

Montreal OPP Forum 3 (Assessment)

The goal of the third forum was to explore and recommend options for assessing orphan drugs for funding by drug plans (public and private) by drawing upon international experience and diverse stakeholders. There was no disagreement that drug funding decisions should include societal values (preferences and ethical considerations) but there was no clear agreement on method for including these criteria. Dr. Stuart MacLeod raised of how to conduct clinical trials with small patient populations, assuring fairness, and incorporating societal values. He posed the Orphan Drug Dilemma as the struggle between optimizing societal resources by not funding rare disorders and the moral obligation to help those most in need. Dr. Edmund Jessop from the UK Department of Health presented the National Specialist Commissioning Advisory Group (NSCAG) which funds services to those with ultra-rare disorders through an elegant process: after experts determine the patient characteristics that would make it likely that someone would benefit from a specific therapy, all patients fitting the criteria are given a chance to receive the therapy. Target outcomes, or benchmarks of effectiveness, are pre-defined and the patient's response is monitored

against these measures. Therapy is continued for those patients who achieved the defined benchmarks and discontinued (after sufficient time) for those who do not. Asked how patient families react when the patient is taken off therapy, Dr. Jessop responded, "We've never had to take a patient off therapy. They take themselves off when it is clear that it is not working."

Professor Michael Drummond concluded with a reminder that health technology assessment was never meant to focus exclusively on economic factors but was intended to include societal values. He called for a "gathering of experts" to determine HTA appropriate to drugs for rare disorders.

National Conference on Orphan Products Policy April 24-25, 2006 (Ottawa)

The objectives of this national forum with international speakers were to raise awareness of Canada's lack of action for patients with rare disorders and to propose options for assessing and funding orphan products, based on the best international experience.

Stakeholders and representatives from several countries met with Canadian patients, researchers, and industry representatives to discuss initiatives to promote development of orphan drugs and access for patients. Dr. Marlene Haffner, former head of the US FDA Orphan Drug Program, reported on the progress achieved in stimulating the development of drugs for rare disorders since the US Orphan Drug Act in 1983, followed by similar legislation in Japan and the European Union, which includes grants for research and development, tax incentives, and market exclusivity.

Dr. Ségolène Aymé of Orphanet / Rare Diseases Task Force (Paris) stated that Europe has a coherent policy regarding rare diseases despite the difficulties raised by 27 different health care systems. Treatment is the ultimate goal and support to research and development is the way forward. Initiatives include a database of clinical trials, website for research projects, and promotion of dialogue among stakeholders through working groups, forums, and task forces.

Sonja van Weely of the Dutch Steering Committee on Orphan Drugs reported that the

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Netherlands had created a national fund specifically to provide access to treatment for patients with rare disorders. Moreover, the steering committee included patient representatives. Overall, she concluded, government or hospitals pay for all prescribed orphan drugs; however, the approval procedure for reimbursement may take some time before the first patient is treated. The Dutch system allocates about 24 million Euros per year to treatment of rare disorders, which is sufficient to provide reasonable access to orphan drugs. However, as new ODs are introduced, the budget will need to be increased.

Dr. Michele Lipucci di Paola, representing the European Organization for Rare Disorders, spoke about the lessons learned from seven years of orphan drug legislation in the European Union. There have been 34 new drugs approved for market. Most new applications are for very rare disorders and most are being developed by small and medium-sized enterprises. There has been tremendous benefit to the centralization of the regulatory and approval process, with support for clinical trials; however, access by patients is still decided by each country and this results in disparities across nations. Eurordis plays an important role in educating and supporting patients and engaging with industry and countries to promote access and availability to affordable medicines for all patients with rare conditions.

Presentations from Canadian patients, government representatives (national and provincial levels), and industry brought the discussion to the next steps for Canada to meet the challenge of innovation and access to treatment for rare disorders.

Chance for Life Fund

The conference concluded with Durhane Wong-Rieger, President of CODR, presenting the highlights from the Canadian Organization for Rare Disorders call for the establishment of a "Chance for Life" Fund to provide fair access to treatment for patients with rare disorders. The proposal asks the governments to establish a dedicated fund equivalent to 2% of public drug funding, or \$200 million per year. An advisory body of experts in rare disorders, including patients would be established to manage the fund. Moreover, experts for each specific disorder would establish the criteria for access to an innovative therapy, with predefined effectiveness criteria and benchmarks. All patients who met the criteria would be eligible for treatment, upon prescription from their specialist, and each patient would be registered, monitored, and assessed against the desired outcomes and potential adverse effects. The Chance for Life Fund would assure monitored access to therapy that is safe, effective, and fair to all patients. All stakeholders are asked to actively participate with CODR and all of the patient organizations to promote the program to the governments and policy decision makers.



One in ten Canadians has been diagnosed with a rare disorder. CODR works with governments, researchers, patients, clinicians and industry to promote research, diagnosis, treatment and services for all rare disorders in Canada. CODR provides information, connects families to one another and supports rare disorder groups and organizations across the country.

"A Chance for Life Fund"

Canada is one of the only developed countries in the world that does not have an orphan drug policy. Not only does Canada not support research and development into treatments for rare and neglected disorders, Canadian patients with rare disorders often do not have the same access to life-saving therapies as patients in other countries. CODR requests immediate action on the following proposals.

- **Establish a national (federal/provincial/territorial) "Chance for Life Fund" equivalent to 2% of the total annual public drug expenditure to be designated for therapies for rare disorders.**
- Establish a multi-stakeholder Advisory Body, including treaters (medical caregivers) and patients, to recommend treatment access for life-threatening or serious rare disorders based on scientific standards and social values (humanitarian, ethical and compassionate criteria).
- Establish Centers of Reference for specific rare disorders, comprised of national and international experts, who will develop criteria for treating patients based on scientific evidence and patient impact and provide on-going surveillance into the real-world safety and effectiveness of these treatments on individual and group basis.
- Provide incentives through Orphan Drug Regulation and policy equivalent to those in the United States and European Union to assure Canadian organizations and researchers are motivated to conduct research and development into treatments for rare and neglected disorders.
- Ensure internationally accepted standards for conduct of clinical trials in rare disorders appropriate for the challenges inherent to very small patient populations (i.e. low number of individuals affected, limited long term data, lack of validated measures, etc.).
- Ensure Health Canada's progressive licensing framework provides appropriate support to the design of clinical trials for very small patient populations and appropriate review of evidence submitted from these trials.

To read more about "A Chance for Life Fund", please visit our website www.raredisorders.ca

Meeting of patients with Morquio disease in Quebec

On Saturday October 20th, 2007, the first meeting of Quebec Morquio patients was held in Laval, Quebec. Eighteen patients attended with family members. The main objective of the meeting was to allow patients and the parents of young patients to meet with Dr Shunji Tomatsu from Saint-Louis University (Saint-Louis, Missouri). Dr Tomatsu is one of only a few specialists working on Morquio disease (mucopolysaccharidosis Type IV or MPS IV) across the world. Dr Tomatsu presented a clinical overview of MPS IV and the status of his research on a potential treatment. Dr Tomatsu has already developed a mouse model for Morquio type IV A and tested a recombinant molecule to be used as an enzyme replacement therapy. The first human clinical trials should begin in 2008 in collaboration with the Swiss-based company, Inotech. Dr Tomatsu also presented the International Morquio Project which comprises an International Registry for Morquio Disease (via the International Morquio Organization). A presentation was also made by Gail Ouellette, Ph.D., geneticist and genetic counsellor, on the genetics of Morquio disease. And, Judy Byrne from the Canadian MPS Society invited the participants to Vancouver in June 2008 for the International Symposium on MPS and Related Diseases. This meeting was a great opportunity for patients and parents to meet and exchange about their experiences in living with this rare disorder.

Gail Ouellette

Réunion de patients atteints de la maladie de Morquio au Québec

Une première réunion de patients québécois atteints de la maladie de Morquio a eu lieu samedi le 20 octobre à Laval. Dix-huit patients et membres de familles ont participé. Le premier objectif de la rencontre était de permettre aux patients et aux parents de jeunes patients de rencontrer le Dr Shunji Tomatsu de l'Université de Saint-Louis (Saint-Louis, Missouri). Le Dr Tomatsu est l'un de quelques spécialistes seulement à travers le monde qui travaillent sur la maladie de Morquio (mucopolysaccharidose

type IV ou MPS IV). Il a fait une présentation sur tous les aspects de la maladie et sur l'état de sa recherche pour trouver un traitement potentiel. Il a déjà développé un modèle de souris pour la maladie de Morquio IV A et a testé une molécule recombinante qui pourra éventuellement être utilisée comme thérapie de remplacement enzymatique. Les premiers essais cliniques chez l'humain devraient débuter en 2008 en collaboration avec la compagnie Inotech qui est basée en Suisse. Le Dr Tomatsu a aussi présenté le Projet International sur la maladie de Morquio qui comprend un Registre international de la maladie (via le "International Morquio Organization"). Une présentation a aussi été faite par Gail Ouellette, Ph.D., généticienne et conseillère en génétique sur la maladie de Morquio. De plus, Judy Byrne du "Canadian MPS Society" a invité les participants à se rendre à Vancouver en juin 2008 pour le Symposium international sur les MPS et maladies associées. Cette première réunion a été une occasion pour les patients et les parents de se rencontrer et d'échanger sur leurs expériences personnelles en rapport avec cette maladie rare.

Gail Ouellette



Meeting of Morquio patients and families in Laval, Quebec, Oct. 2007



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Canadian Fabry Association: Update

Much has been accomplished since the last Canadian Fabry Association General Meeting held in May 2006 in Halifax, Nova Scotia where the CFA was officially established and the first slate of Board of Directors elected. At that meeting, there were two government officials who clearly stated that access to enzyme replacement therapy (ERT) was imminent. We all know now that was not the case!

The Executive Committee and Board of CFA met on several occasions since May 2006 with the priority being access and funding of ERT. It has been somewhat frustrating but significant progress has been made. However many challenges remain! It has been a long long road since the adhoc group of patients and supporters first started working together back in August 2003. The CFA worked with many in a process to access ERT & focused on inequalities of patients with rare disorders. We sent over 2000 letters to Health Ministers, used media exposure and were able to secure many meetings and campaigns.

Access to ERT in Canada today is very restrictive and limited. We do now have a limited 3 Year Funding Agreement between Prov/Fed/Mfg which was approved in August 2006. In addition we now have a separate Canadian Fabry Registry called the Canadian Fabry Disease Initiative (CFDI) which started only in October 2006. The CFDI provides limited access to ERT with Canadian made criteria which are the most stringent in the world. Because there are so few Fabry patients in Canada on ERT (75 now with

plans up to 150) we believe that this study will not prove anything. In fact in 3 years from now the CFDI will provide data that we already had in the international community three years ago. The CFA's position remains that Canada should administer ERT for Fabry as a preventive therapy prior to organ failure, in line with the international community.

We are currently involved currently conducting a survey of Canadians with a lysosomal storage disorder (Gaucher's, Fabry's MPS, Pompe's and other). We are asking about their experience with healthcare services, including diagnosis, access to drug therapy, including enzyme replacement therapy, access to other healthcare services and psycho-social support for them and their family. We will summarize the findings to help make the case for the quality of care for patients with lysosomal storage disorders, and what else is needed. Our plan to present the poster for the Lysosomal Disease Network World Symposium scheduled for February 13-15, 2008.

The aim and goal of CFA continues to be to find and diagnose Fabry patients in Canada and lead them to proper treatment. With the help and support of many, the CFA will continue to grow, develop and network with other organizations in an effort to improve the lives of Fabry patients and others suffering from other life threatening rare disorders as well as their families and care givers in Canada and around the world. Fabry patients and their families require a comprehensive care program that fits their particular needs.

Adrian (Ed) Koning

Canada Takes Part in 1st International Rare Disease Day February 29, 2008

It is not too late to take part in an international event to raise awareness about rare diseases and disorders. CORD will be hosting a number of national and provincial activities: to raise awareness and to call for action to implement the Chance for Life Fund and improve the quality of life for all persons affected by rare disorders.

CORD invites you to join us for a rally at the Parliament in Ottawa on February 28, 2008 and special workshops and celebrations on February 29th.

This is a unique opportunity that will happen only once every four years on February 29th. If you are affected by a rare disorder, if you are part of a patient organization, or if you just care about improving the lives of those with rare disorders, CORD invites you to join us. CORD is looking for enthusiastic, energized, creative volunteers to help plan and carry out activities in each province and local community.

Potential Activities:

- Rally on Parliament in Ottawa on February 28th
- Rally at Provincial Legislatures
- National Rare Disorder Photo Contest
- Fundraising Activities
- Newspaper Insert
- Media Events
- Public Service Announcements

Please volunteer by sending an email or leaving a phone message with your contact information and any specific ideas or just say "I want to help." Send to: info@raredisorders.ca or call toll free : 1-877-302-7273.

International Rare Disease Day



February 29, 2008

"A rare day for very special people"

Canadian Association of Pompe Disease 2nd Annual Meeting

The second annual meeting of the Canadian Association of Pompe (CAP) was held in Toronto on November 17 and 18, 2007. The meeting brought together Pompe patients, family members, caregivers and health professionals. The first day of the meeting was an educational day. Robin Casey, MD (Inherited Metabolic Disorders Program, Alberta Children's Hospital, Calgary), presented an overview of Pompe disease including the treatment options that are available or are in development. Joanne Koskie (Cohn & Wolfe) gave some tips on how a patient organization can advocate its needs and demands towards the government. Margery Konan and Dorothy Creaser, leaders of the workshop "A practical approach to living better – learning to manage the challenge of a chronic disease" offered by the Institute for Optimizing Health, presented the approach used in these workshops and went through some parts of this interactive workshop with the meeting participants. Finally, Ralph Kern, MD (Genzyme Canada) gave an update on the current status of Myozyme in Canada. On the second day, Judy Byrne from the Canadian MPS Society gave an overview of their organization's activities, challenges and suc-

cesses and offered CAP many suggestions for advocacy and fundraising. The rest of the meeting was an opportunity for the CAP members to exchange what they accomplished during the last year in each of their provinces and to establish priorities for the upcoming year. The two main priorities are, first, to reach more Canadian Pompe patients (last year's founding meeting brought together seven patients from across Canada with this rare disorder; since last year, five other patients were reached by the Association). Second, to continue advocacy for access to Myozyme, the only available approved treatment for Pompe disease. In June 2007, the Common Drug Review recommended that Myozyme be listed only for infantile-onset Pompe patients (with symptoms before 12 months of age). CAP will continue to advocate for access and funding for all Pompe patients. Ian MacPherson, vice-president of CAP, presented a video he produced himself and posted on YouTube.com (**S.O.S. - Help Make a Difference in Ian's Life** <http://fr.youtube.com/watch?v=PuLOiPIfGW0>).

Gail Ouellette



CAP 2nd Annual Meeting, Nov. 2007 held in Toronto

Featured Board Members: Ann Gloyn and Joseph Witalis

Ann Gloyn of Brantford, Ontario has been a member of CORD's Board of Directors since 2004. She has an active and healthy family with two children and husband involved in travel sports.

Ann is an Education Specialist Teacher, who has worked with the Ministry of Education for Ontario in a Provincial residential school for Blind, Visually Impaired and Deafblind since 1985. She holds undergraduate degrees in physical and health education, a degree in psychology, and in education. She holds additional qualifications for teachers in Deafblind Education, Blind Education and Special Education. She also has done some post graduate work. Ann has presented at National and International conferences and workshops, and has lectured in University courses related to additional disabilities and sensory loss.

Ann has been active in volunteer work serving four years on the Board of Directors for "Canadian Deafblind and Rubella Association, Ontario Chapter"; served on a local community advisory committee for disability issues; serves on the Professional Advisory and Research committee for the CHARGE Syndrome Foundation in the U.S.; and has been a Board member since 2002 of "CHARGE Syndrome Canada", a National registered charity dedicated to providing information and resources and support research on the leading cause of deafblindness in infants and children.

Joseph Witalis of Toronto, Ontario has been a member of CORD's Board of Directors since September 2005. Joseph is a long-term survivor of a rare cancer of the bone marrow – multiple myeloma - and in this connection has been an active volunteer in numerous health care organizations; since 1994, he has volunteered as a Community Representative with Cancer Care Ontario's Research Advisory Committee and the Cancer Care Ontario Regional Council for South-East Ontario; the National Cancer Institute of Canada's Terry Fox New Initiatives grants panel; the Peterborough District Health Unit Task Force on Complementary and Alternative Treatments; and served as Chair of the Autologous Blood and Marrow Transplant (ABMT) Peer Support Group at Princess Margaret Hospital in Toronto. Joseph continues to be a volunteer member of the Princess Margaret Hospital Patient Education Committee.

Joseph worked for thirty-two years in the management of residential mental health centers for behaviour-disordered and mentally ill children and adolescents and their families until experiencing the disabling combination of cancer and its treatment, a very highly toxic clinical trial in intensification of stem-cell transplantation in 1996. Since shortly after that treatment, Joseph has left employment and focused his energies on health care advocacy.

Carrying the burden of care with orphan disease: A patient's perspective

The burden for accessing appropriate treatment and care, usually falls to the patient when they have a rare or orphan disease. This has certainly been my own experience and the experience of many others.

When I was diagnosed with Waldenstrom's Macroglobulinemia, a rare blood cancer, I quickly found out you cannot rely on just any hematologist to give you the best advice. At the appointment with the 'specialist', I was given a diagnosis and a prescription in one fell swoop. It was only after doing my own investigative research, mostly on the internet that I found out the prescription could wait. On the internet I found out about the International Waldenstrom's Foundation and was connected to a local support group. In fact, at the time it was the only support group in Canada. How I managed to find a support group for a rare cancer in my own vicinity was quite amazing.

In 2002, I was diagnosed with WM – an indolent lymphoplasmacytic lymphoma, with an overall incidence of 3 to 5 per million (10,000 in the USA and Canada). Because WM is rare, most hematologists may never see a patient who has this disorder. And unlike most cancers, the desired approach for treatment is 'watch and wait' until symptoms have a negative impact on overall quality of life, or become life-threatening. In my own case, I was finally forced into treatment because of a serious pleural effusion. Physicians tend to look at what works in indolent non Hodgkin's lymphoma and use similar approaches on WM.

At a CORD forum in October 2005, I reported that after having 8 months of Rituxan + cvp (cyclophosphamide, vincristine and prednisone) I obtained a complete remission, but WM is not curable by drugs and my disease will come back. In my case, being a good responder, it was recommended by a WM expert in the US that I go on maintenance rituxan every three months for a period of two years to extend the response period. That means I am given a single infusion of rituxan every three months. Research in follicular lymphoma demonstrates that maintenance rituxan not only extends the response period, but increases overall survival.

The difficulty for WM patients is there have been no clinical trials specifically to test maintenance therapy in WM. And like many other rare disorders, the numbers are too small to make it cost-effective. At a recent International WM conference in KOS, there was a unanimous recommendation by WM experts to push for and prioritize a clinical trial with MR. In the USA, maintenance rituxan is funded under the umbrella of NHL (non-Hodgkins lymphoma). In Ontario, WM is considered 'distinct' from NHL. My oncologist at Princess Margaret Hospital initially refused to prescribe MR because it was not approved, but primarily because it was not funded.

Evidence based medicine is great if you happen to get a brand-name disease and the doctors who treat patients are familiar with the treatments and protocols, however when you are faced with a rare disease, it can be extremely frustrating. Out of necessity, patients must learn to become an 'expert' in their own disease and thus are often more knowledgeable than the doctors who treat them. Patients are often required to advocate for themselves to fight for a treatment at a time when their own health and stamina is compromised.

In my case, I had two options. I could go to a private clinic in Ontario at my own expense. The quote was approximately \$43,000 and about half were personal non-insured costs. The other choice was to go to Roswell Park Cancer Institute in Buffalo because the non-insured costs were considerably less. In other provinces such as B.C. and the Maritimes, maintenance

rituxan is approved based on the recommendation of the patient's hematologist. And patients who get this treatment do not have to pay for it themselves.

At this point I contacted Durhane Wong-Rieger, President of CORD. She encouraged me to begin writing letters to the Minister of Health and Long-Term Care et.al. and tell my story at a CORD forum.

I feel strongly that educating physicians about WM is key, so I was able to make some connections to bring a renowned WM expert to Toronto to speak to physicians at Sunnybrook and PMH. Inadvertently, through this effort, I found out about a rituxan assistance program offered in Canada through the drug company – Roche. Once my doctor signed the forms, I would be able to get the infusion costs covered in Ontario at one of the new Bayshore Clinics and my own private insurance would cover the drug costs. However, due to delays, I was forced to go to Roswell Park in Buffalo for my first treatment. Then I was able to get my next two treatments through the Bayshore Clinics – the first one in London, then the second one in Toronto.

Finally, maintenance rituxan was approved in Ontario, however, I was told it would not apply to me because the initial treatment protocol had to be completed after July 2006 and I finished in May. I spent a lot of time on the phone with Cancer Care Ontario over that one. Then I was informed I would be able to get MR through the hospital because I had been approved through an extended 'compassionate program'. I was in! So, in June, I had my first fully approved (and funded) treatment at PMH. At the beginning of September I was due for another treatment, however when I checked to confirm the time, I was told that, because the papers had not been sent in, the hospital had not been reimbursed for the June treatment and I was denied further MR treatments. I had to get back to CCO for support again. It took a while before the paperwork issue was resolved and I was back on track. To date I have had 5 out of the 8 treatments and am due for another one in early December.

Getting a rare disease is a steep learning curve. It is imperative that you become an active partner in your own medical care and you can't do that unless you really understand your disease. If you have been told that your cancer is 'rare' it usually means that your doctor has not treated many of these cancers. It is essential that patients of rare diseases locate a 'centre of excellence' and get a second or third opinion, if necessary. You need to find a doctor or an institution that is focused and experienced with your disease. These are the places that are engaged in exciting, state of the art research and are having the best results with this disease. As Hamilton Jordan points out in his book, *Never a Bad Day*, "you don't want to be the patient that your doctor(s) go to school on!" And if you go for that second opinion, it is imperative that your local oncologist is open, and confident enough to listen to the 'expert's opinion and advice. And, as Jordan also points out, "there are harsh realities of the economics of health care." This is part of the equation in the decision making process regarding treatment. In my experience, I felt it was necessary to find the best medical advice regardless of the cost – regardless of what my insurance covered. Then and only then could I decide whether or not to go down that road – whether or not I could afford to act on that advice.

Betty McPhee

First-ever drug therapy for PKU, a rare disorder What about timely access for Canadian patients?

My son, Toronto resident and university student, has become a consumer of American health care to get access to a breakthrough drug therapy for a rare disorder. Our journey began when my son was diagnosed 20 years ago, thanks to newborn screening, with a brain-threatening metabolic condition called phenylketonuria (PKU). PKU is a genetic disorder affecting approximately 50,000 patients worldwide. It is caused by a deficiency of the enzyme phenylalanine hydroxylase (PAH), which is required for the metabolism of the amino acid phenylalanine (Phe), found in most protein-containing foods. If the enzyme is insufficient, Phe accumulates in the blood and brain, resulting in complications including severe mental retardation and brain damage, mental illness, seizures and tremors, and cognitive problems.

Until this year, the only treatment for PKU patients is a vile-tasting medication and a highly restrictive diet (virtually no meat, fish, nuts

or legumes) that most patients fail to maintain to the extent needed to control blood Phe levels. As PKU patients on diet mature, there is growing evidence of nutrition issues due to lack of minerals and trace elements. Ontario Health pays 100% of the costs of an approved list of prescription drugs, formulas and specialty foods medically necessary for metabolic patients. Other provinces provide less support. Recently, Kuvan, a breakthrough drug, has done well in clinical trials. US FDA approval was granted this past month.

At my son's urging and with the assistance of Dr. Annette Feigenbaum of Toronto's Hospital for Sick Children, we managed to enroll him in a special access program through Children's Memorial Hospital in Chicago. Because not all PKU patients respond to Kuvan, the first step was to determine response. Luckily, my son responded very well very quickly, going from a baseline of 824 micromoles of Phe per litre blood to

210 mmoles/l after 15 days. Now we start to experiment with how much natural protein he can handle.

Kuvan is not yet available in Canada, and there's the rub. The American company, BioMarin, which has been co-developing Kuvan for PKU, is providing the drug under the special access program at no cost to patients. This access program will end two months after FDA marketing approval and BioMarin expects to be paid at that point. They will be applying for Health Canada approval. If they are granted fast-track status, that would mean a decision in nine months, considerably after the end of the access program in the US.

We are actively pursuing all available options to continue Kuvan treatment for my son without interruptions.

John Adams



SHARE YOUR PHOTO!

In celebration of the 1st International Rare Disease Day, the Canadian Organization for Rare Disorders invites all members of the Rare Disease/Rare Disorder community to share your story and to share your photo.



2008 CORD Photo Contest

Do you have a favorite photo that shows the spirit, love, courage, or challenge of living with a rare disorder? Send us a digital copy and you could be a winner in the 2008 CORD Celebration of Persons with Rare Disorders! Contest will be judged by a panel of esteemed jurors. Winning photos will be published in CORD's national newspaper feature supplement on February 29, 2008.

Four categories of photos:

- Children affected by a rare disease/disorder
- Adult affected by rare disease/disorder
- Family or group (those directly affected and those not)
- Activity involving persons affected by rare disorder (recreational, arts, medical, etc.)

Deadline: January 31, 2008.

Name of submitter, names of featured persons, and contact information must be included.

Please email your photo entries to: info@raredisorders.ca

2008 CORD Story Contest

Do you have a personal story that helps illustrate what it means to live with a rare disease or disorder? It can be inspiring, funny, poignant, sad, or even scary. Stories may be about an individual, family, your favorite doctor, or something that happened to you.

Stories must be between 250 and 500 words in length and must be submitted electronically. The winning stories will be published in CORD's national newspaper feature supplement on February 29, 2008.

Suggested themes: overcoming personal challenges, support of family and friends, challenges with health care, starting a patient group, working with a doctor, teacher, or therapist.

Deadline: January 31, 2008.

Name of submitter, names of featured persons, and contact information must be included.

Please email your story entries to: info@raredisorders.ca

WANTED: Corporate Sponsors for Photo and Story Contests.

Please visit www.raredisorders.ca for more information.



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