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## PRESIDENT'S REPORT

The fiscal year 2005-06 was a year of significant challenge and opportunity for the Canadian Organization for Rare Disorders. It was also a year of significant transition, which saw CORD expand from its hometown roots of Coaldale, Alberta to a new corporate location in Toronto, Ontario.



CORD President,  
Durhane Wong-Rieger

At the Annual General Meeting on September 27, 2005 held in Newmarket, Ontario, a new Board of Directors was elected. One of its first mandates was to rationalize the organizational structure, including specification of job descriptions and the roles and responsibilities of Board and staff. Over the course of the next two months, the Board reorganized and stabilized the officers. Durhane Wong-Rieger, president of the Anemia Institute was elected president; Helen Tsenekos moved into the position of past-president; Adrian (Ed) Koning, chair of the Canadian Fabry Society, remained as vice-president; Ann Gloyn, vice-president of Charge Syndrome Canada assumed the responsibilities of secretary; and John Adams assumed the role of treasurer. The other members of the Board were Kirsten Harkins, Executive Director with the Canadian Society for Mucopolysaccharide and Related Diseases and Joseph Witalis. Kim Craig was elected in November.

In November 2005, CORD hosted another successful casino with Alberta Gaming Commission. CORD continued to support the efforts of patients and patient organizations to gain access to new therapeutic agents, including those for Fabry patients, those with MPS-I, as well as a new treatment for Gaucher's Disease. CORD continued to play a key role in calling for increased newborn screening in all provinces. While Ontario has made significant strides, the other provinces have yet to respond to the need for early identification of those at risk for genetically linked disorders. And the organization began planning around the collaboration for and promotion of the Orphan Drug Policy document prepared in 2005.

In February and March 2006, CORD went through a difficult challenge from some members, resulting in a struggle to define the direction and leadership of the organization. A special members meeting held on April 8, 2006 confirmed the Board of Directors and the officers of the organization. CORD has now re-established a head office in Toronto, Ontario with an Alberta office in Edmonton. Since the move, CORD has taken the lead, in collaboration with the Canadian Genetic Diseases Network, BIOTECANADA, and Health Canada in planning for the first Canadian Conference on Rare Disorders and Orphan Products Policy. The conference, scheduled for April 23-24, 2007 in Montreal, Quebec will serve as the culmination for a series of forums to be held during fall 2006 and winter 2007. In 2006, CORD also initiated the Corporate Leaders Forum, following the model of the (USA) National Organization for Rare Disorders Corporate Council and EURORDIS Corporate Roundtable. Membership is open to all corporations providing research and development for rare disorders. Finally, CORD has reinstated a drive for affiliate (patient group) members.

We look forward to a very productive 2006-07.

Durhane Wong-Rieger, PhD, President

# Forum I: 'Options for Funding Drugs for Rare Disorders under Ontario's New Drug Legislation'

The first of three forums initiated by CORD in an effort toward developing an Orphan Drug Policy in Canada was held in Toronto on October 24, 2006. The conference entitled—'Options for Funding Drugs for Rare Disorders under Ontario's New Drug Legislation' was well attended by several pharmaceutical companies, patient groups and a representative of the Ontario Ministry of Health.

The objective of the conference was not only to review the impact of Bill 102 in Ontario for Ontario patients with rare disorders but also to review patient access to drugs for rare disorders within the Canadian and international contexts. The longer term goal is to develop recommendations to the government so that Canada may join other countries which already have an Orphan Drug Policy.

The ODP must allow patients with rare life threatening disorders in Canada the same access to drugs and medications available to other patients in other parts of the world.

The good news is that Health Ministers in their interim report on a National Pharmaceutical Strategy agree that the issue they call "Expensive Drugs for Rare Disorders" has to be addressed.

Several speakers, patients and company representatives presented their own case studies demonstrating why the current review process via

the Common Drug Review is not working and must be changed.

All stakeholders in attendance at the meeting agreed that an ODP would be a step in the right direction.

A highlight of the meeting was one guest speaker, Dr. Andreas Laupacis, the former Chair of the Canadian Expert Drug Advisory Committee (CEDAC) the group that the Common Drug Review (CDR) relies on for expert opinion and evaluation on any new drug considered for funding. He spoke on 'The Canadian review mechanisms and applications to drugs for rare disorders and the CDR/CEDAC experience' and agrees that changes are required to the current process. He admitted that the issue of rare drugs are societal, policy decisions and whether we as a society want to treat these drugs and patients differently because of the special issues in dealing with rare disorders. He said that the CDR process should be opened up and made more transparent. Having a well defined "orphan drug policy" definitely makes a difference in other countries and makes it easier to manage the issues around rare or orphan drugs. The UK Citizen's Council is a successful model, where transparency has been achieved, and patients and citizens are actually players. In the UK, rare conditions are funded nationally through NSCAG. In the Nether-

lands, there is a dedicated committee to evaluate drugs for rare diseases and make funding recommendations. In Germany, funding for one new drug discussed was approved simultaneously with marketing approval.

In the US, there are public and private payers reimbursing orphan drugs.

Canada is far behind other countries in orphan drug policy. There is a need to move forward to determine the guiding principles for addressing these problems, and to look at models for possible solutions.

Subsequent Forums on topics such as 'Appropriate methodologies for R&D into rare disorders', and 'Reimbursement and Funding Issues' are scheduled.

One objective is to engage the Ontario Drug Strategy Secretariat as well as others from provincial Health Ministries at the next sessions. It would also be very helpful to bring in other decision makers to have a better chance for dialogue.

CORD is seeking help in organizing future forums: finding speakers and coordinating logistics.

\*please note that full proceedings Forum I are available on our website.

Adrian (Ed) Koning



MEETS



## Learning from the First (and still the Best)

Since becoming president of the Canadian Organization for Rare Disorders, one of my ambitions has been to visit the place where it all began, the National Organization for Rare Disorders. This is the rare disorders' equivalent of the trek to Mecca, and my goal was not only to glean wisdom from their years of experience in serving and advocating for the rare disorders community but also to gain inspiration in moving CORD forward. What can we do in Canada to promote collaboration and to mobilize the rare disorders community to achieve the level of respect and political clout that NORD has earned in the United States and internationally? When I asked Dr. Stephen Groft, Director of the NIH Office of Rare Disorders how the US Congress happened to create this Office that clearly recognizes the need for a specific focus on research and support for rare disorders, he responded without hesitancy, "The same reason they passed the Orphan Drug Act in 1983; it was all due to NORD and their getting pa-

tients, families, and patient groups to advocate together."

We have all benefited from the Orphan Drug Act in the USA, which has led to the development of 300 drugs for rare disorders over the past two decades. NORD was the impetus for the European Organization for Rare Disorders (EURORDIS), which also championed the passage of the European Orphan Drug Act in 2000 and keeps rare disorders in the forefront of healthcare policy in all European countries. I had met Jean Campbell, Vice President of Development and Diane Dorman, Vice President for Public Policy at various forums and was delighted at the warm response to my request for a visit this past August.

What a smart operation! Housed in its own building in the very small town of Danbury, Connecticut, NORD is conveniently located just minutes from New York City, within driving distance of some of the major pharmaceutical and biologics developers and manufacturers, and a few hours by

(Continued on page 3)

train from Washington DC.

I had the privilege of sitting down with Abbey Meyers, the founder and president, and it was immediately apparent why NORD has been so effective. Abbey offered a wealth of political insights as well as a few simple axioms for success: make allies, work relentless, and surround yourself with dedicated and talented staff. NORD offers a model that is worth emulating.

The NORD Corporate Council brings together leaders of the orphan disease community "to promote the interchange of information among voluntary health agencies, health-related industries and regulatory agencies, and to facilitate dialogue about research and development of treatments for rare diseases." Under the leadership of Jean Campbell, the Corporate Council has flourished from its initial dozen members to an active group of 40+ corporations that meet on a regular basis with NORD patient groups, researchers, and government. Mary Dunkle oversees NORD's extensive information services including a rare disease database, resource library and newsletter, as well as information support.

A unique and very valuable service for patients is the compassionate access program for orphan treatments coordinated by Maria Hardin at NORD on behalf of various companies. One very important member of the team not in Danbury is Diane Dorman, Vice-president for Public Policy, who is located in Washington DC. I look forward to NORD's future collaborations with NORD and will certainly take up their very generous offer to support NORD in its efforts toward establishing an Orphan Products Policy in Canada and increasing NORD's services to the rare disorders community.

## **NORD 2006 Annual Conference: Road Map for Rare Disease Research**

Following this visit, I was delighted to attend NORD's 2006 Annual Conference, Road Map for Rare Disease Research. Held in Washington DC at the end of September, the purpose was for the rare disease community to learn "how rare diseases fit into scientific research being conducted at the National Institutes" and to hear directly from the experts about important research issues. The agenda included key representatives from the US Food and Drug Administration and National Institute for Health. It was my first experience attending a NORD conference and I was particularly interested in learning the level of engagement of patient representatives with researchers and



**Durhane pictured here with NORD President, Abbey Meyers**

government representatives.

The topics were challenging, focused on the future direction of research, not just what research was being done but new ways in carrying research that would provide the more effective translation of research ideas into innovative treatments for patients.

Key was the NIH Road Map and the promotion of biomedical research towards applied problems and useful knowledge. Patient groups were extolled to take a role in helping to define problems and to participate in the design and implementation of clinical trials. Similarly, the US Food and Drug Administration (the regulatory sector) spoke of its newest initiative, the FDA Critical Path. Dr. Janet Woodcock of the FDA was followed by Dr. Raymond Woosley of the nonprofit C-Path Initiative in presenting this new approach to education and research. The goals of the Critical Path are threefold: change drug development process to provide faster access to drugs, develop programs to ensure safer drugs and safer use of drugs, and create educational programs to enhance pharmaceutical development.

Two of the most interesting panels provided a "behind the scenes" look at areas not usually accessible to patients. One presented the drug approval process with members of industry and the FDA Office of Orphan Products Development. The second was a simulated Institutional Review Board (IRB) meeting, which included a re-enactment that invited audience participation in challenging the research proposal under review.

The acting FDA Commissioner, Dr. Andrew

Von Eschenbach, was on hand to talk about the important role of patients in clinical trials as well as the translation of basic science into applied diagnostic and treatment products. Two perennial government favorites (!) were Dr. Marlene Haffner, Director of the FDA Office of Orphan Products Development, speaking on the FDA's initiatives, Dr. Stephen Groft, Director of the NIH Office of Rare Disorders, whose included discussion of a pilot program to promote new genetic test development and better understanding of the rare disease.

Most impressive was the tremendous amount and of patient participation and the level of discourse. It was obvious that years of active engagement have created a large contingency of informed and capable patient advocates representing rare disorders, and their involvement was not only welcomed by government and industry but also considered as invaluable. There is much that Canada, NORD, and the rare disorders community can learn and should directly imitate from our very successful counterparts to the south. There has been a tremendous culture shift towards the importance and value of treatment and support for rare disorders in the USA (and Europe). The Canadian rare disorders community must not be left behind and hope that we will benefit from the windfall of initiatives and innovations pioneered in other countries.

Durhane Wong-Rieger, PhD

## CORD at Mayfest

*An awareness event sponsored by The Ontario Association of the Deaf*

Canadian Organization for Rare Disorders (CORD) hosted an information table during "MAYFEST", at St. Lawrence Market in Toronto. It was a well-attended event, with considerable numbers in the hundreds passing through information tables which lined each side of the building. CORD volunteer Stephen Greathead as well as CORD board member Ann Gloyn, gave out pamphlets to help bring CORD to the attention of participants, mostly of whom were hearing impaired. There are rare disorders which cause hearing impairment and deafness. One individual who approached our table was interested in promoting an awareness of "Waardenburg syndrome". This is a rare genetic congenital disorder, which causes effects which range in severity. Clinical features can include not only hearing impairment or deafness, but facial abnormalities, and abnormalities in pigmentation, which can not only be the skin but the iris. Both eyes can also be of different colours. The genetics of this syndrome is interesting, much like the findings in Usher's syndrome, in that there are a number of types of Waardenburg, which groups unique sets of symptoms with gene findings. One of these includes a diges-

tive gastrointestinal disorder, or "Hirschsprung disease". What was interesting is that most of the population who were older did not know their etiologic or specific causes of deafness. Many just reported that "...its genetic, I don't know". Some of the younger population seemed to have similar clinical features to a newly reported and under diagnosed syndrome "CHARGE syndrome", but were unaware of it. CORD needs to help provide information to individuals so that they are aware of the full implications. That could include future information on any possible treatment, availability of clinical trials, research studies, therapy and approaches, and future affects on families. One other opinion must be mentioned, which is a strong tribute to "deaf culture". Individuals who are deaf do not consider being deaf or hearing impaired a "disorder", attempted to promote a change of name for "Canadian Organization for Rare Disorders". Their desire was to reflect something in a name which considers it a "gift of genetics" to be different and unique.

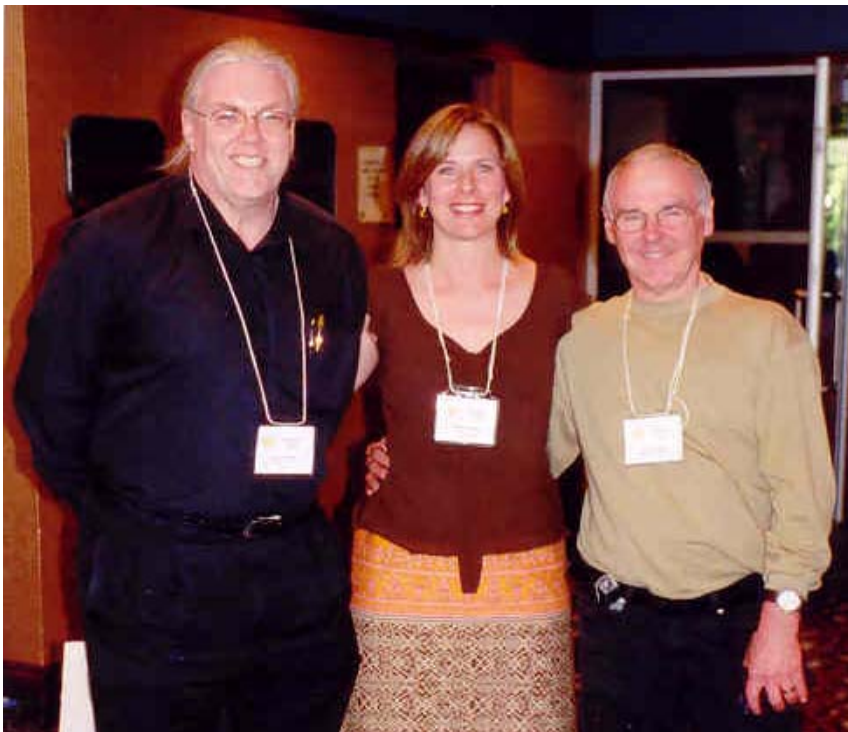
Ann Gloyn

## The Canadian MPS Society Invades Blue Mountain!

From July 14 to 16, 2006, the Canadian MPS Society held their Family Conference at the Blue Mountain Resort in Collingwood, Ontario. The conference was focused around sessions which people had appreciated in the past, and add some new elements which would improve the experience for everyone. In addition, for the first time in many years, we had a number of MPS adult patients in attendance. With the advent of new therapies, this will be an element that will become increasingly prevalent at our future conferences. Some of the speakers that presented were – Dr. Joe Clarke, Dr. Beverly Antle and Lindsay Moir. Dr. Clarke presented the much appreciated overview of MPS and related diseases. Dr. Clarke's years of experience are an invaluable resource for the Society and we always appreciate his contributions. Dr. Antle again provided her insight into issues of grief, but also enlightened us with the aspects of hope that permeate our lives and how hope can help us in our journey with MPS kids. Again, Lindsay Moir provided some exceptional insight into the education system and how it accommodates our kids. We were very pleased this year to have a few new speakers. Dr. Martin Collis of Vancouver, Canada gave a wonderful introductory talk on Healing, Humor and High—Level Wellness. His infectious and positive outlook on life was a great way to start the day. For example, he had some great acronyms. He introduced us to "T.R.U.S.T.": tomorrow's results ultimately start today". His most memorable message to us was that we should live our life "M.E.L.L.O.W.": with Magic of mind, Exercise, Laughter, Love, Optimal nutrition, and Wonder.

Another new speaker this year was Durhane Wong-Rieger, President of the Canadian Organization for Rare Disorders. She enlightened us on the steps that must be taken to have a drug approved and funded in Canada. This of course is of tremendous importance to us as new therapies become available. Finally, one of the highlights of our conference was having Dr. Ed. Wraith of Manchester Children's Hospital in attendance. Dr. Wraith is probably one of the most experienced MPS doctors in the world, not only in a clinical setting, but also due to his involvement with clinical trials of the new therapies. He gave a great talk on his experience with current treatments, and gave an outline of the new research and therapies which we may expect in the future. While we know that research takes years to get to the clinic, it is very encouraging to see that people are working on treatment and cures for our kids.

Judy Burne  
Canadian MPS Society



(L-R) Dr. Ed Wraith, Kirsten Harkins, and Dr. Joe Clarke at the 2006 MPS Family

# CORD/OCMR Weekly MediaScan

The Canadian Organization for Rare Disorders / Organisation Canadienne des Maladies Rares is pleased to be able to provide a new service to the rare disorders communities. A weekly sampling of news from around the world having a bearing on rare disorders has been available since August of 2006.

This service, called the *CORD / OCMR MediaScan* is available electronically to members and affiliate organizations. It consists of links to current articles in the press, radio, and television sources from around the world. These links are identified by theme, meaning the relevant disorders or areas of interest; such as newborn screening, social policy, and technical announcements. It is sent out weekly on Mondays, and also includes examples from an archive of earlier articles published in earlier months. The date of the article, as well as the source, author and country of origin are included so that interested members of the rare disorders community can access information of interest to them.

The articles are stored on the *CORD / OCMR* website and will be uploaded weekly. This will help to ensure the information posted will be accessible by anyone who wishes information on articles on rare disorders. We compile the news links, but since the actual articles are provided by other news sources we can not ensure that they will be always in use or 'live'. For this reason we will be sure to have the most up-to-date *MediaScan* posted on our website. A few of the articles may require the

viewer to become a subscriber to the news media, though we most often try to avoid any inconvenience in accessing them.

Articles are available in English when published in English, and in French when published in French. As a national organization, we feel it is important to provide access to both founding languages of Canada. Until we acquire professional translation services, we will not be able to provide total bilingual information.

In the future, we would like to be able to develop an in-house .pdf file archive of all the articles included in the *MediaScan* to be made available to persons wishing to conduct research into various aspects of communication of information relevant to rare disorders.

We also will soon be developing a searchable database of articles, so those seeking information in the news about specific disorders, specific policies, or specific countries, will be able to find articles about specific disorders, policies, and countries. This could be a valuable tool for advocacy in Canada and in other jurisdictions.

Of course, help is always appreciated. If you would like to volunteer some time to help in making the *MediaScan* an even more complete resource for the rare disorders communities, please contact *CORD / OCMR* at [info@raredisorders.ca](mailto:info@raredisorders.ca).

Joseph Witalis  
Chair, Resource Development Committee

## Newborn Screening for Rare Disorders

A modern medical miracle exists to protect the health of Canada's 340,000 newborn babies each year from a small but growing list of the thousands of rare disorders and the good news is that most of Canada's provincial health programs are expanding these early detection services.

The miracle is called newborn screening. It is designed to detect disorders with proven treatments but which are difficult or impossible to diagnose before irreversible harm occurs. It involves taking a tiny blood sample for every newborn baby, shipping each sample to specialized labs with sophisticated technologies to analyze key biomarkers and short-term and long-term follow-up including diagnosis, treatment, parent, professional and public education and program evaluation. Most Canadians are shocked to realize newborn screening in most provinces used to rank as the weakest in North America. For example, Mississippi is a very poor jurisdiction but it screens its babies for 57 rare disorders, while Ontario and New Brunswick screened their babies for only 3 disorders, meaning those two provinces were tied for last in Canada and the United States.

Ontario is expanding its newborn screening from 3 to 28 disorders by the end of 2006. In November 2006 Ontario became first province to screen all newborns for sickle cell disease and just announced plans to add cystic fibrosis by the end of 2007. When Ontario completes this expansion it will be the first province to screen newborns for all 29 core disorders recommended by the American College of Medical Genetics. Congratulations Ontario.

This fall, Alberta announced plans to expand its screening from six to 19 and Alberta is the first in Canada to include cystic fibrosis. The Alberta expansion is to be complete by April 2007. Congratulations Alberta. The Alberta expansion does not yet include sickle cell disease, for example. Saskatchewan has been a leader in Canada, screening babies for 36 disor-

ders and adding two more this year. It has not yet announced plans to include cystic fibrosis or sickle cell disease, for example.

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Advisory Committee of the Seed of Life Philanthropic Organization, a new charity dedicated to the prevention of sickle cell disease.

Front (L-R): John Adams CORD Board Member; Moni Williams; Lan Tunji-Ajayi, founder of SOLPO; Dr. Audley James; Revivaltime Tabernacle of Toronto.

Back (L-R) Timothy Tunji-Ajayi, Dr. O.F. Alisafe, Dr. Isaac Odame, leading pediatric oncologist/hematologist and founder of the Hemoglobinopathy Group of Ontario.

There continue to be great disparities in newborn screening across Canada. Taking into account announced plans to expand; newborn screening scope ranges from a high of 38 disorders in Saskatchewan to a low of 5 in British Columbia and 6 in Manitoba and Newfoundland & Labrador. No US state screens for a few as 6; 40 screen for 25 or more. Newborns in the Northwest Territories, Nunavut and Yukon are screened by adjacent provincial programs.

For more information on newborn screening please visit our website [www.raredisorders.ca](http://www.raredisorders.ca). In the resources section we provide a very detailed information chart regarding which disorders are currently being screened for each province.

John Adams  
CORD Treasurer and NBS Advocate

Number of Disorders Screened by Province (as of November 25, 2006)		
Newfoundland & Labrador	6	- 2 added in 2006
Prince Edward Island	15	- 3 added in 2006
Nova Scotia	15	- 3 added in 2006
New Brunswick	3	- 12 to be added in 2007
Quebec	15	- expansion under review (plus 11 transport disorders from a unique urine sampling program)
Ontario	24	- plus 4 by end 2006 and 1 by end 2007
Manitoba	6	- expansion under review for 2007
Saskatchewan	38	
Alberta	6	- plus 13 by April 2007
British Columbia	5	

## Canadian Fabry Association Update – Access to Enzyme Replacement

The Canadian Fabry Association (CFA) was incorporated as a non profit organization to raise awareness and educate the public about Fabry disease, a very rare life threatening, genetic lysosomal storage disorder. Patients are missing a key enzyme which causes extreme pain, inability to sweat, gastrointestinal problems and eventually kidney failure, heart attacks and strokes. The average life span of a male patient without treatment is 40-50 years of age.

The CFA also encourages and support research into the causes, treatment and management of Fabry disease, but most importantly to arrange for, co-ordinate and increase the facilities available for diagnosis, consultation and treatment of those suffering from Fabry disease. The diagnosis of Fabry disease like any other rare disorder is the first and most critical and frustrating challenge for any one with a rare disorder. You can not treat what you do not know.

The good news is that about 6 years ago, enzyme replacement therapy (ERT) was approved to treat those with Fabry but at a cost of approximately \$250,000 per patient per year. But not in Canada. Canadians with Fabry disease continue to struggle with access to ERT and the CFA has advocated for several years that the publicly funded health care system in Canada should fund and provide ERT to those that require it.

Health Canada approved ERT for use in Canada back in January 2004, but the provinces refused to cover it until August 2006 when a three year funding agreement between all provinces, the federal government and companies was approved. The challenge now is that the Canadian Research Protocol (a Canadian made registry) has not yet

been approved. Everyone who was on treatment via a clinical trial or compassionate use is now back on it which includes those in Ontario and Nova Scotia many who were cut off for over a year. The bad news is that those who are “new patients” and meet the strict Canadian guidelines have not yet started ERT.

CFA together with CORD and one other organization co-authored a letter to all health ministers urging that ERT be implemented immediately (a commitment they made back in October 2005) and to express the concern at their suggestion that the Fabry agreement be used as a model for a policy on Expensive Drugs for Rare Disorders, as discussed in their June 2006 Progress Report on the National Pharmaceutical Strategy. Canada should join the rest of the world by participating in the appropriate international patient registries.

Canada is far behind the rest of the world in providing ERT to its Fabry patients as well as providing access to rare or ‘Orphan’ drugs. In fact Canada is the only developed country in the world not to have an Orphan Drug Policy. This is one reason that the CFA has joined the Fabry Intentional Network (FIN). The primary aim of the FIN is to facilitate collaboration between organizations to support those affected by Fabry disease. It seeks to do this primarily through enabling communication, promoting best practice and acting as an independent forum for Fabry patients around the world.

FIN is connected to over 20 countries including Canada and the USA. FIN was incorporated in the Netherlands utilizing the Dutch laws for non profit organizations. Membership is free and open

to any national patient organization in which Fabry patients are represented. A board of directors was elected and the current President and Treasurer are both from the Netherlands. The past president of Norway’s Fabry Association is the Vice President and the Secretary is Adrian Koning, a Fabry patient, and President of the Canadian Fabry Association. His wife Marlene is also a board member and has a passion for helping caregivers of Fabry patients.

The board meets regularly via telephone conference. Canada and Poland currently need the most help as these countries do not fund or provide access to enzyme replacement therapy (ERT) based on international criteria and guidelines. To improve communication one focus is on developing its website. There are two companies that manufacture ERT and although competition is good, this fact has caused strife within the Fabry community. The principle foundation of FIN is to be neutral and independent in all of its communication, action, and decisions worldwide and one long term goal to promote a single world wide International Fabry Conference that allows a place where all relevant aspects and information of Fabry disease and current or future drugs and therapies can be shared in a non biased environment.

If you would like more information about the CFA, FIN or the situation of access to rare disorders in Canada, contact Adrian Koning at [koning@interbaun.com](mailto:koning@interbaun.com)

Ed (Adrian) Koning  
Vice-President, CORD

# GENETIC ALLIANCE ANNUAL CONFERENCE 2006

The “Genetic Alliance 2006 Annual Conference” in Washington celebrated 20 years of Excellence in Advocacy. The focus was to build bridges, develop partnerships, and applaud the inspirational leadership of all groups who support or give information. “Genetic Alliance” assists mostly groups of individuals and families who are living with genetic conditions in the United States, and helps them to achieve their mission. “Genetic Alliance” is able to leverage voices with similar needs, by collecting the groups together and build capacity. “Genetic Alliance” has provisions in place to be able to affect research development and funding; help individuals gain access to services; assist groups in partnership development; and encourage and support emerging technologies.

Individuals who may have genetic conditions face the same issues in the United States and Canada. There are many barriers to seeking health care services which address their often rare disorder; there is a great delay in diagnosis; a lack of information both in access and in treatment options. In Canada we face another critical roadblock: a consistent Federal approach to these issues is needed, and Canadian politicians need to develop through consultation from stakeholder groups an “Orphan Drug Policy”.

In opening sessions on “Leadership in Action”, we learned that for organizations committed to their mandate, there needs to be four elements of sustainability. These are fundraising and resource development; marketing and communications; networking and collaboration; and volunteer development. With a clearly defined mission in place, needs must then be identified and then programs or services built around this. Throughout this process, collaboration with other organizations is essential. It was suggested that personal stories of those who have genetic conditions be documented, and groups must “tell their story” in the same way frequently. Once a marketing mindset is in place, and a compelling case for support has been developed, the funding base must be diversified to be successful. When donor's are interested in an area, the work the organization does should match this interest, build relationships with supporters, and show good stewardship of resources it does have.

Worksheets were given as tools to assess what organizations were doing, in order to identify strengths. For further “break-out” sessions, groups were then matched for strengths and weaknesses, so everyone could learn from each other. Other

sessions were research oriented, demonstrating examples of putting “research into action”. Some issues which were covered included large-scale screening; comparative genomics, and other applications in Genetics. Company representatives from “Gene DX” uncovered their “tools of the trade”. Other sessions demonstrated how partnerships are being developed in the Industry sector, both in education and testing collaboration.

The “oversight of genetic testing” was interesting in that genetic testing is playing an increasing role in healthcare delivery, and tests can be used for the basis for profound medical decisions. However, there does exist questions as to the accuracy of some tests. Who regulates testing, and whether there is “pre-market” approval of genetic tests are areas yet must be touched on in legislation. Examples from a variety of groups demonstrated how the education plan for the genetic disorder had been developed. In these sessions, uniform strategies were presented in order to “get the word out” about the condition.

Other sessions worked with parents in order that they develop their own family history trees, identify risk factors, and be their own advocates to talk with healthcare providers. There are a variety of impacts on families, and the transition to adult programs for the individual with a genetic condition was a huge area of concern for many families. Problems exist for families, such as finding expert medical assistance needed once children reach adulthood; finding and funding services, especially assistance with vocational issues. Parents have to be key advocates in this process when their children leave the education system. There was a suggestion developed that groups work together in order to advocate for “adult neurodevelopmental clinics” and “adult genetic centers”, where networked relationships can be developed and evaluation and care can be coordinated. While this conference held many other sessions and topics, there was a focus on other useful areas such as ethical and cultural issues; newborn screening; models for chronic care; and especially highlighted resources and toolkits. The greatest learning was from meeting those who had an interest in a rare disorder or genetic condition, or who suffered from one. The variety of conditions which do exist was an incredible education process in itself!

Ann L. Gloyn  
Education Specialist



## CORD Participates in Jeans for Genes Day 2006

Did you know that 3 of 5 Canadians will experience a disease with a genetic component in their lifetime?

Jeans for Genes Day is the annual fundraising campaign for the Canadian Gene Cure Foundation. Participating organizations encourage their employees & students to wear jeans to show support for the cause - finding cures for genetic diseases. For each \$5 donation, participants receive a Jeans for Genes denim button and an entry into a national draw to win great prizes, such as a shopping spree or a trip to Whistler. All money raised goes to support Canadian research into treatments and cures for genetic diseases in children.

CORD and the Institute for Optimizing Health Outcomes participated in this year's Jeans for Genes Day fundraiser on Thursday, October 12, 2006 by hosting an informational booth in the lobby of the office building at 151 Bloor St. West in Toronto. Starbucks generously donated the coffee, and staff and volunteers offered their time to greet people and explain the importance of research into genetic diseases. We raised over \$450 for CGCF and raised public awareness at the same time - and had fun doing it!

To participate in the next Jeans for Genes Day, check the website [www.jeansforgenes.ca](http://www.jeansforgenes.ca) for information. You can become a 'corporate genie' or a 'school genie' to organize fundraising activities in your community.

1st Canadian Conference on  
Rare Disorders and Orphan  
Products Policy

April 24-25, 2007  
Crown Plaza Hotel  
Ottawa, Ontario

# National Deafblind Conference – Needs of those with Rare Disorders

Recently I attended the “National Deafblind Conference” in Winnipeg, Canada. There was a demographic study just completed which showed that 3,306 persons living in Canada are “deafblind”. Like with every rare disorder, this number greatly underestimates the number of actual individuals which do exist. There is difficulty in reaching those who are considered “deafblind”, especially when they live in nursing homes. There is also the potential for lack of clear diagnosis and identification. In school age population, children who have incomplete vision and hearing loss can be missed, especially when other disabilities are present.

For individuals who have combined vision and hearing loss to such an extent that it interferes with their ability to gain undistorted information from the environment and communicate can be considered “deafblind”. It is known to be a functional definition. The population cannot be compared with each other, because each reason for the sensory loss, and the degree of loss will differ in each individual. There are also many with additional disabilities, which complicates a complete picture of what a “deafblind person” really is..

In each area of Canada, one still hears arguments over who meets the criteria, and who does

not. For many, a “deafblind” label will help them get some services to function more independent in society, which includes having more depth in their existence as they struggle to make sense out of the partially distorted world. “Deafblind” does not necessarily mean total loss of vision and hearing, and that is another reason why looking to find these individuals in Canada is a complicated process. Beside trying to identify actual population which does exist, the “Study of Deaf-Blind Demographics and Services in Canada” held focus groups across the country which helped identify needs. A summary of these is listed in a report at [www.cdsdb.ca](http://www.cdsdb.ca). It should be noted that because numbers have been identified, it does not mean that further work can be done in helping these individuals meet their needs. All information is confidential, and so potential “numbers” can be the only result of this study.

As the “Canadian Organization for Rare Disorders” works toward promoting an understanding for Canadians of the need for an “Orphan Drug Policy”, we need to look at some of the needs of the population of those who are “deafblind” which could be included. In other countries, an “Orphan Drug Policy” is more of a complete document, which goes beyond the need

for the implementation of rules regarding certain drug therapy and clinical trials.

For example, in Manitoba there is no “assistive devices program” like there is in Ontario. Many who are “deafblind” have limited financial resources, and the need for devices to help them with daily living is evident. Those who are not in educational programs or in workplaces are at a particular risk in not having simple tools to make their life easier. Those who are in private nursing homes are again of particular risk. There needs to be a consistent federal approach to funding for these devices.

Secondly there is a need of increased access to training and assistive device technology. Useful technology has improved in the past ten years, and will continue to do so. A federal program which does ensure these individuals who are “deafblind” not only have the greatest opportunity for the most recent technology, but have the training to make it most beneficial to their lives to live independently, should be included in a national “Orphan Drug Policy”. With increased independence, the “pay-back” can be huge, and these people have the potential again to become contributing members of society.

Ann Gloyn

## 1st Canadian Conference on Rare Disorders and Orphan Products Policy

First Canadian Conference on Rare Disorders and Orphan Drug Policy

April 24 & 25, 2007 Crown Plaza Hotel Ottawa

**WHAT:** “Made-in-Canada” solution to research, development and access to “orphan products” for rare diseases

**WHO:** Canadian and international researchers and clinicians, biotech and pharmaceutical developers and manufacturers, patients and patient organizations, policy makers, economists, and healthcare funders

**WHY:** Unlike other countries, Canada has no “orphan products” policy that specifically supports development and access to drugs and devices for diagnosis and treatment of rare disorders.

**HOW:** Multi-stakeholder consensus conference drawing upon international and Canadian experience to address incentives, access programs, and patient partnerships

**Hosted by:** Canadian Organization for Rare Disorders  
Canadian Genetics Disease Network  
Biotech Canada

**For information updates, visit:** [www.raredisorders.ca](http://www.raredisorders.ca)  
or call: 1-877-302-7273



# Canadian RARE DISORDERS AND ORPHAN PRODUCTS POLICY

## Agenda 2007

### Forum 2

#### Increasing Canada's Productivity to Meet Unmet Needs for Rare Disorders

February 6<sup>th</sup>, 2007

Renaissance Vancouver Hotel Harbourside

Registration Fee: \$50

The purpose of the forum is to help define a Canadian environment that will support researchers and industries that want to contribute productively by translating research to useable products to help meet the needs of those with rare disorders (prevention, identification, diagnosis, and treatment). The forum will present profiles of leading Canadian companies and products in development, Canadian researchers engaged in translation for market applications, and the patient organizations participating in support of clinical trials and product development. Additional topics for discussion include: federal and provincial support and incentives for orphan product development, Health Canada's regulatory process for pre- to post-market investigation, and international collaboration.

The format will include expert presentations, panel discussions, and work sessions. The outcomes of the forum shall include recommendations for Canadian policies and programs to support development of therapies for rare disorders (to be discussed at the Canadian Conference on Rare Disorders: Therapy Development and Access) as well as the articulation of an ethical framework for industry-patient group partnering and identification of opportunities for collaboration.

\* Please note that proceedings from Forum 1 are available on our website.

### Forum 3

#### Canadians with Rare Disorders—Assuring Access to “State of the Art” Diagnoses and Therapies

March 20<sup>th</sup>, 2007

Montreal, Le Saint Sulpice

Registration Fee: \$50

The goal of this forum is to develop recommendations that would assure Canadians with rare disorders have appropriate access to “state of the art” therapies and diagnostic techniques, based on international best practices within an environment that promotes safe, effective, monitored use. The forum will present examples of access strategies, policies, and programs for diagnosis and treatment of rare disorders that are common across jurisdictions as well as unique programs, such as those funded through hospitals, centers of excellence, and “special access envelopes.” Presentations will include the Special Access Programme, Common Drug Review and Canadian Expert Drug Advisory Committee, National Pharmaceutical Strategy, Quebec Conseil du médicament, Ontario Drug Strategy and other provincial plans. Finally, the agenda will address the role of patients and public in drug access decisions.

The outcomes include recommendations for Canadian policies and programs to assure patient access to innovative therapies and diagnostic tests for rare disorders (to be proposed at Canadian Conference on Rare Disorders: Therapy Development and Access). The outcomes will also be used to prepare input to the National Pharmaceutical Strategy on the priority issue of innovative therapies for rare disorders.

## First Canadian Conference on Rare Disorders and Orphan Drug Policy

April 24 & 25, 2007 Crown Plaza Hotel Ottawa

Registration Fee: \$100

The goal of the conference is to achieve a consensus toward a “made-in-Canada” solution to promoting the development of and access to novel drug therapies for patients with rare disorders.

This solution should address government policies that will make Canada an attractive place for researchers and companies to identify and bring new therapies to market for rare and ultra rare conditions, based on the experience with incentives and development support available in the USA, Europe, Japan, and other countries.

The solution should also address the optimal procedures for access to rare therapies by Canadian patients, including factors such as: design of clinical trials, agreement on surrogate markers and endpoints, requirements for post-market surveillance, cost-effectiveness assessment, quality of life and ethical considerations, international best practices, and the parameters for conditional, special, or compassionate access.

Finally, the solution should address other issues that would support optimal use of drugs, such as new born screening and early diagnosis, patient registries, genetic counselling, professional and public awareness and education, and chronic disease management, including self-management.

### Forum Participation

Invitation to participate in all forums is extended to researchers and clinicians working in rare disorders, to healthcare decision makers at all levels of government and institutions, and to corporations who are members of the CORD Corporate Leaders Forum. Participation is also open to all affiliate (patient group) members of CORD. Individual CORD members may attend on “space-available” basis.

All sponsoring organizations are invited to send delegates to the Conference, based on level of sponsorship. All patient group and individual members of CORD are invited to attend. To register, please contact CORD directly at 416-969-7464 or [info@raredisorders.ca](mailto:info@raredisorders.ca)

