



# Alpha-1 Canada Community News

January 2011

What's New at  
[www.alpha1canada.ca](http://www.alpha1canada.ca)

- Ontario withdraws funding for augmentation therapy, join the fight to reinstate
- Podcast with Durhane Wong-Reiger, PhD on the development and implementation of a Canada-wide regulatory framework for rare disorders

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## Another year of making a difference in the lives of Alphas

by Mimi McPhedran

As 2011 begins, we can't help but look back on the most impactful event of 2010; that being the devastating news that The Ontario Drug Benefit program of the Ontario Ministry of Health and Long-term Care had ceased approving funding of alpha-1 antitrypsin augmentation therapy (Prolastin®) for newly prescribed patients.

There is still much work to be done on this front. However, if the information from our December drop-in support meeting with Dr. Durhane Wong-Reiger is any indication, the good news is that Health Canada has recognized and is in the process of acknowledging rare diseases.

Reflecting back and planning for the future is also a New Year's exercise. It's a time to look toward new horizons and develop new strategies, to rediscover strengths and gear up for new challenges. Here at Alpha-1 Canada we have been doing exactly that.

The Board of Director's is proud to announce that we have been awarded a grant from the Ontario Trillium Foundation for a special project to develop on-line support groups. The short version summary is that we are in the process of creating a model for internet-based peer support groups that will provide health benefits for people who are suffering with rare diseases. The long version is that

this extensive project will take well over a year to complete its many phases. We will certainly keep you abreast with more details as time progresses.

Since this project is so time consuming and has a mandated completion date our Board of Directors made a decision to reduce our monthly signature newsletters to a quarterly publication. It is our goal and that of our staff to maintain the same quality support and services during this project stage. Never fear, Jim and Vanessa remain dedicated to providing you with e-blast communiqués as news breaks.

The New Year is also a time for connecting with family, building community and engaging in new resolutions. Please make participation in Alpha-1 Canada your top resolution priority.

Here are some simple, quick and easy things you can start immediately. Pick a few or do them all!

- ◆ Pay it forward. Let others know what you have discovered about your disease.
- ◆ Increase awareness in your community and most importantly with your families by sending our newsletters to everyone in your e-mail list and bring a copy to your health practitioners. Also, we have a quantity of brochures explaining our dis-
- ◆ ease available to you and your physician.
- ◆ Discuss blood serum level testing with your siblings.
- ◆ Encourage your family members and physicians to register with Alpha-1 Canada.
- ◆ Let us know what you would like to read in our newsletters and listen to in our drop in support group meetings.
- ◆ When attending funerals, where charitable donations are requested in lieu of flowers, please make a donation to Alpha-1 Canada. We will ensure the deceased's family will be informed of your donation and a tax receipt will be provided to you.
- ◆ Engage in regular active gifts of kindness.
- ◆ Donate to Alpha-1 Canada whenever possible no matter the amount.
- ◆ Help us help you!
- ◆ Call us or drop us an e-mail just to say hello or tell us your story.

Yours is the voice we want to hear. And we are looking forward to hearing from you.

Wishing you a Healthy & Happy New Year!

## Closing the Distance: Patient Support in the 21st Century

by Jim Mundy

A few months ago we told you that we had received a grant from the Ontario Trillium Foundation (OTF) for a project related to support groups. The project is called, "Closing the Distance: Patient Support in the 21st Century" and aims to bring patients with rare disorders, their families and caregivers closer to others in similar circumstances over the Information Highway.

We applied for this funding in collaboration with the Canadian Organization for Rare Disorders (CORD).

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The health benefits of peer support groups for people with significant disease are well documented. In fact research has shown that people who take part in support groups have lower levels of anxiety and depression and live longer than those who do not take part in such activities. But rare disease sufferers are sparsely scattered, making face-to-face peer group meetings impractical.

In the case of more common diseases like cancer or diabetes, the pool of potential group members is large and creating a group in a town or even part of town that can meet regularly and

in person is a very real possibility.

The same cannot be said in the case of rare diseases. Any given rare disease sufferer may be the only person in town with that disease or the only person for miles. This used to mean that support groups for people with rare diseases were not possible or required hours of driving. Depending on a patient's symptoms, driving may have been out of the question, or made the meeting such an undertaking that they were scheduled far apart and attended infrequently.

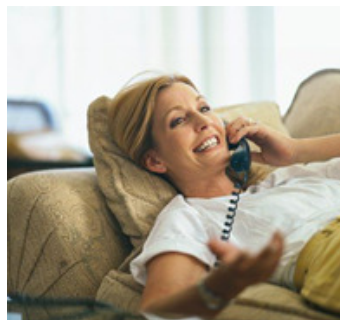
This OTF funding is allowing us to develop an on-line model for support groups specifically designed using Internet tools to bridge those miles and make Support Group attendance possible and easy for the majority of people with a rare disease like alpha-1 antitrypsin deficiency.

Once developed and implemented for Alpha-1 Canada, a generic version of this model will be made available for use by other rare disease groups through CORD.

Our model is a systematic manual and method for virtual (internet-based) peer support groups allowing all rare disease sufferers and their caregivers to experience these critical and essential health benefits.

The manual and leader training materials are almost complete and in the next month we will begin a process of recruiting and training support group leaders, and shortly thereafter, recruiting support group members. We hope to be able to attract enough vol-

unteer leaders to establish at least one on-line support group per province although some provinces may not have enough Alphas and need to combine with another nearby province and other provinces will be able to accommodate more than one.



Whether you are interested in leading one of these groups, or being a member, this is a wonderful opportunity to meet fellow Alphas, inform them about the tricks you have learned, share some good conversation and contribute to a project that will benefit rare disease patients across Canada.

Please get involved.

Think about joining one of these groups and, ideally, being a leader.

We will provide further details in upcoming communications, but if you just can't wait to say yes, please contact Jim Mundy at jim.mundy@alpha1canada.ca or 1-888-669-4583.

### Tip of the Month

What spikes your blood glucose (sugar levels) more: A sweet potato or brown rice?

Surprise – it's the brown rice!

To read full article: [http://www.morethanmedication.ca/en/article/index/glycemic\\_index](http://www.morethanmedication.ca/en/article/index/glycemic_index).

### PODCAST

Listen to our latest podcast with Durhane Wong-Rieger, PhD on the development and implementation of a Canada-wide regulatory framework for rare disorders : [http://www.alpha1canada.ca/Educational\\_Podcasts](http://www.alpha1canada.ca/Educational_Podcasts)

## Donations

We would like to take this opportunity to send out a hearty 'Thank you' to all of you who donated during 2010, including the generous donations we received during the holiday season.

Your donations throughout the year are used to support a number of programs and services for Canadians suffering from Alpha-1 Antitrypsin Deficiency and their caregivers, such as the patient support hotline, the bilingual newsletter and website, and many more.

If you are considering making a donation, we gladly accept a cheque or money order made payable to "Alpha-1 Canada" or by credit card securely through CanadaHelps.org.

THANK YOU!

## CORD's Arctic Quest

by Vanessa McLaughlin

It was at the beginning of October while attending the Canadian Organization for Rare Disorders' (CORD) 3rd Annual Conference, "Many Rare Diseases – One Common Voice" that I first heard of a fundraising initiative called Quest for the Arctic Circle: Summer 2011.

This extraordinary quest to show that people with rare disorders can do just about anything with the right treatment and support is now recruiting one dozen enthusiastic individuals. You don't need to have a rare disease to make the voyage. CORD welcomes family, friends, and supporters. Although the trip does not require any previous experience and you will be hiking with experienced guides, you should be fit and capable of carrying up to 15 kg (35 lb.) approxi-



mately 8 km a day. Voyagers will depart from Ottawa and fly to Baffin Island on August 13th to return on August 21st.

Alpha-1 Canada is working toward having an Alpha involved in the Quest, but anyone who is able to raise the required funds is wel-

come to participate. We will keep you posted on this development and also encourage you to learn more about the many ways you can be part of this unique challenge by visiting:

<http://raredisorders.ca/ArcticQuest2011.html> .

## Interesting Research

*The following are summaries, known as abstracts, of recent studies of Alpha-1 and COPD. Because of copyright law we can usually only provide abstracts.*

*Please note that Dr. DW Cox (pictured below), who is a member of Alpha-1 Canada's Medical Advisory Board, is a co-author of the study in the first abstract*

*and that the full version of that article can be read here: <http://www.ncbi.nlm.nih.gov/books/NBK1519/>.*



### Alpha1-Antitrypsin Deficiency.

Gene Reviews [Internet]. In Pagon RA, Bird TC, Dolan CR, Stephens K, editors. Seattle (WA): University of Washington, Seattle; 1993-2006 Oct 27 [updated 2008 Feb 06].

Schlade-Bartusiak K, Cox DW.

### Excerpt

**Disease characteristics:** Alpha1-antitrypsin deficiency ( $\alpha$ 1ATD, AATD) caused by homozygosity for the common deficiency allele, PI\*Z, is characterized by chronic obstructive pulmonary disease (COPD) in adults and liver disease in children and adults. COPD, specifically emphysema, is

the most common manifestation of AATD.

Smoking is the major factor influencing the course of COPD. The onset of respiratory disease in smokers with AATD is between age 40 and 50 years; in non-smokers, the onset can be delayed to the sixth decade. Non-smokers often have a normal life span.

Although reported, emphysema in children with AATD is extremely rare. AATD-associated liver disease, present in only a small portion of affected children, is manifest as obstructive jaundice and raised serum aminotransferase levels in the early days and months of life. The incidence of

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## Interesting Research

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liver disease increases with age; liver disease in adults, manifest as cirrhosis and fibrosis, is not necessarily associated with a history of neonatal liver disease. Hepatocellular carcinoma (HCC) has been reported. Clinical disease is infrequent in heterozygotes, except in some smokers.

**Diagnosis/testing:** The diagnosis of AATD relies on demonstration of low plasma concentration of alpha1-antitrypsin (AAT) and either observation of a deficient variant of the protein AAT by protease inhibitor (PI) typing or detection of mutations in both copies of the gene SERPINA1, which encodes AAT. PI\*Z (resulting from the mutation p.E342K) is the most common deficiency allele. Ninety-five percent of AATD results from the presence of two Z alleles. Molecular genetic testing is clinically available.

**Management:** Intravenous augmentation therapy (regular infusion of purified human AAT to augment deficient ATT serum concentrations) has been recommended for affected individuals whose FEV1 is 35%-50% of predicted and who have quit smoking yet continue to show rapid decline in FEV1 despite optimal medical therapy; however, appropriately controlled trials have not been carried out. Avoidance of smoking (both personal and passive), occupations with exposure to environmental pollutants, and exposure to mineral dust, gas, and fumes is recommended.

Liver transplantation, the preferred surgical treatment for advanced liver disease,

can provide a cure because the donor liver produces AAT.

### **Genetic counselling:**

AATD is inherited in an autosomal recessive manner. When both parents are heterozygotes, each sib of an affected individual has a 25% chance of being affected, a 50% chance of being a carrier, and a 25% chance of being unaffected and not a carrier. In the rare instance in which one parent is homozygous (PI ZZ) and one parent is heterozygous, the risk to each sib of being affected is 50%. Unless an individual with AATD has children with a reproductive partner who is affected or a carrier, his/her offspring will be obligate heterozygotes (carriers) for the disease-causing mutation. Carrier testing is available on a clinical basis by PI typing (isoelectric focusing) or mutation analysis for sibs and offspring of affected individuals. Prenatal diagnosis for pregnancies at increased risk is possible by molecular genetic testing once the diagnosis has been confirmed in an affected family member.

### **Cardiovascular and musculoskeletal co-morbidities in patients with alpha 1 antitrypsin deficiency.**

Respiratory Research. 2010 Dec 7; 11:173.

Duckers JM, Shale DJ, Stockley RA, Gale NS, Evans BA, Cockcroft JR, Bolton CE. Section of Respiratory Medicine, Wales Heart Research Institute, School of Medicine, Cardiff University, Heath Park, Cardiff, UK.

### **Abstract**

**BACKGROUND:** Determining the presence and extent

of co-morbidities is fundamental in assessing patients with chronic respiratory disease, where increased cardiovascular risk, presence of osteoporosis and low muscle mass have been recognised in several disease states. We hypothesised that the systemic consequences are evident in a further group of subjects with COPD due to Alpha-1 Antitrypsin Deficiency (A1ATD), yet are currently under-recognised.

**METHODS:** We studied 19 patients with PiZZ A1ATD COPD and 20 age, sex and smoking matched controls, all subjects free from known cardiovascular disease. They underwent spirometry, haemodynamic measurements including aortic pulse wave velocity (aPWV), an independent predictor of cardiovascular risk, dual energy X-ray absorptiometry to determine body composition and bone mineral density.

**RESULTS:** The aPWV was greater in patients: 9.9(2.1) m/s than controls: 8.5(1.6) m/s,  $p = 0.03$ , despite similar mean arterial pressure (MAP). The strongest predictors of aPWV were age, FEV1% predicted and MAP (all  $p < 0.01$ ). Osteoporosis was present in 8/19 patients (2/20 controls) and was previously unsuspected in 7 patients. The fat free mass and bone mineral density were lower in patients than controls ( $p < 0.001$ ).

**CONCLUSIONS:** Patients with A1ATD related COPD have increased aortic stiffness suggesting increased risk of cardiovascular disease and evidence of occult musculoskeletal changes, all likely to contribute hugely to overall morbidity and mortality.

If you know of any research, articles or other publications that would be of interest to our readers please contact us.

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Visit us on the web  
at [alpha1canada.ca](http://alpha1canada.ca)

Our website is continuously updated with useful information for Alphas, their caregivers and healthcare providers, as well as news on promising research. Make a habit of checking our website regularly so you won't miss out on exciting updates and always read our monthly newsletter from top to bottom.

Help us spread awareness by sharing this newsletter with your family and friends.

If you would like to receive this newsletter by e-mail, please contact us at 1-888-669-4583 or [vanessa.mclaughlin@alpha1canada.ca](mailto:vanessa.mclaughlin@alpha1canada.ca)

This newsletter is designed to support, not replace, the relationship that exists between you and your physician. It is not the intention of this newsletter to provide specific medical advice but rather to provide the Canadian Alpha-1 Community with information to better understand their health and their diagnosed disorder.

Specific medical advice will not be provided and Alpha-1 Canada urges you to consult with a qualified physician for diagnosis and for answers to your personal questions.

**Alpha-1 Canada**  
**Making a difference in the lives of Alphas**

### Changes to the Alpha-1 Canada Board of Directors

It is with sadness that we announce that Joanne Wicker has resigned from the Alpha-1 Canada board of directors. The Board extends their sincere thanks and best wishes to Joanne for her many contributions and we look forward to her continued assistance and dedication to the Canadian Alpha-1 community.

Joanne is a mother of four children ages four through 13, one of whom has Alpha-1 antitrypsin deficiency. She is also a children's programmer at their local library in Alberta. A few of

her great joys in life are to make children feel special; to make learning fun, and to connect parents with the information that they need to make good family choices.

Joanne brought a special point of view to the board and has agreed to continue to help with developing programming for Little Alphas and their families.

"I am just too busy with family commitments at this time, and as my family must come first I can see no alternative," Joanne

said, "I feel that I cannot give good service and dedication to the Alpha 1 Canada Board and to my family."

If you think you might be interested in a position on the Alpha-1 Canada board of directors, contact Jim Mundy for more information (1-888-669-4583 or [jim.mundy@alpha1canada.ca](mailto:jim.mundy@alpha1canada.ca)). The board usually meets once each month by evening telephone conference call.