



Alpha-1 Canada Community News



June 2010

What's New at www.alpha1canada.ca

- **New procedure for Alpha-1 testing**
- **2010 Annual General Meeting report from the board of directors**
- **Ontario withdraws funding for augmentation therapy, join the fight to reinstate**

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Alpha-1 Canada welcomes Vanessa McLaughlin

We told you last month that we were in the process of hiring a new staff member at Alpha-1 Canada. Vanessa McLaughlin is now on board and has a message for you.

Hi, my name is Vanessa McLaughlin and I am the new Part Time and Bilingual Administrative Assistant of Alpha-1 Canada. I am very excited about working with Jim Mundy, the Board of Directors, the Medical Advisory Board and you, the Alpha-1 Community. Prior to joining the Alpha-1 team I worked as a teacher, both in Canada and England. I was raised in Québec and am looking forward to helping develop a sense of community from coast to coast for English

and French-speaking Canadians.



I am hopeful that I will be able to carry out the Alpha-1 mission and provide you with information in the monthly newsletter and support on the website. That being said, these resources and all of Alpha-1

Canada's services are for all members of the Canadian Alpha-1 community, so feel welcome to contact me directly with issues, concerns, ideas or suggestions to help make them even more valuable tools for all!

Vanessa

vanessa.mclaughlin@alpha1canada.ca

We are very excited to have Vanessa on the Alpha-1 Canada team and look forward to her efforts and the positive effects they will have on our community.

Please join the Board of Directors in welcoming Vanessa.

US Alpha-1 Conference a huge success

The American Alpha-1 Association hosted its 19th Annual National Education Conference on June 11-13, 2010 in Orlando, FL. A record-breaking 603 attendees, including ten Canadians, gathered at the Hilton Walt Disney Resort to hear the latest on Alpha-1 and

its treatments and to meet other Alphas.

It was a weekend filled with outstanding speakers and lots of activities for families and children to participate in. Alpha-1 Kids provided entertainment and games for the children while the parents

were able to attend the meetings.

You will have the chance to download the presentations shortly at www.alpha1.org. Next year the 20th Annual National Education Conference will be in St. Paul, MN, June 10-12, 2011.

Many Rare Disorders—One Common Voice

The Canadian Organization for Rare Disorders (CORD) Annual Conference on Rare Disorders is our "once-a-year" chance to come together as a rare disorders community to celebrate, share, learn and inspire.

Join CORD on October 1 - 2, 2010 at the Sheraton Ottawa Hotel for CORD 2010 "Many Rare Disorders—One Common Voice!" Check the CORD website (www.raredisorders.ca) for more information on the

conference and travel sub-



Many Rare Disorders — One Common Voice

sides.

Only by working together have we been able to make significant progress toward a Canadian Plan for Rare Disorders. At the conference you will have an opportunity not only to learn about but also to provide input on key initiatives, including

- a 5-year rare disease research competition

- draft regulatory framework for orphan drugs
- access programs for drugs for rare diseases
- proposed Centres of Excellence and Reference, and more.

This year's conference will include more workshop sessions for "hands-on" learning.

Please complete the pre-

conference survey to indicate your preferences for topics and content.

Go to: www.surveymonkey.com/s/H39RND

Further conference details will be available on the CORD website (www.raredisorders.ca) soon.

Please complete the survey by June 30, 2010.

Will Canadian MPs be receptive to national 'orphan-disease' strategy?

Unlike some countries, including the United States, Canada does not have a national strategy to deal with rare disorders. This means that Canadians do not always have timely and affordable access to the best therapies and there is little incentive for pharmaceutical companies to develop new therapies.

During a special session of the parliamentary health committee, Durhane Wong-Rieger, CORD's president highlighted Canada's lack of national strategy.

"It means a more difficult time getting clinical trials

for patients with rare disorders; it means that, unfortunately, Canadian patients with rare disorders are one of the last patients in the developed world to get access to new medicine."

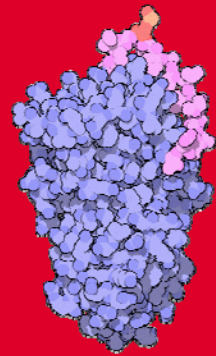
Part of the problem is that there is no official 'orphan status.' "The Canada Health Act is out-of-date and is not keeping up with scientific advances, which is detrimental to all patients, not just ones with rare diseases," said Wong-Rieger.

Health Canada's regulatory regime needs to be updated. For example, the

rules around clinical trials stipulate that a certain number of patients be included. However, the same rules should not apply to a rare disease as it affects so few people.

Even if drugs for rare disorders are successful in getting through the regulatory framework and are approved for use, the next challenge is convincing governments to pay for them. As funding for certain drugs varies from one province to another, access may differ from province to province. "At the end of the day, if you live in one province you may get a lot better access than if you live in other provinces and that of course, is unacceptable," said Wong-Rieger.

While almost everyone agrees that millions of people have benefited from the Orphan Drug Legislation in the United States, some three million Canadians who are affected with a rare disorder will continue to suffer and face an uphill battle until a national strategy is in place.



Alpha-1 Antitrypsin

**Join CORD in
October 2010
for "Many
Rare
Disorders—
One Common
Voice!" in
Ottawa**



**Alpha-1 can affect
infants and
children as well**

Healthy Living Canada Online Self-management Project is now accepting participants into the research study

Healthy Living Canada is the online version of the community-based Chronic Disease Self-Management Program (CDSMP). This is the first time the on-line Healthy Living Canada workshops have been offered in Canada and participants will be recruited from across Canada. The free, online self-management program is only offered in English at this time.

Any adults with any type of long term health condition can participate. You need just 2 hours a week for 6 weeks to complete this workshop online. You can do the workshop anywhere you have access to the Internet and even people

who are learning how to use the computer will be able to take part in this user-friendly workshop.

You will be asked to fill out

Any adults with any type of long term health condition can participate.

three questionnaires for the study: one when you sign up, one in 6 months, and one a year after you have finished the workshop. Each questionnaire takes between 20 to 30 minutes. There will be about 25 people with chronic conditions in the workshop so you will be able connect with others

and share new ways to address your health condition. You will use a screen name during the course and your personal information is always protected.

You will be given a free copy of the book *Living a Healthy Life with Chronic Conditions* when you start the workshop.

Register online at www.healthylivingcanada.org or if you have more questions, please email Learn-More@healthylivingcanada.org

This Canada-wide project is sponsored by the Government of Alberta and Alberta Health Services.

Alpha-1 Drop-in Support Group

It's for you



Is pain, fatigue, stress or other symptoms from your ongoing health condition stopping you from living your life to the fullest?



This Canada-wide pilot project is sponsored by the Government of Alberta and Alberta Health Services.

Government
of Alberta

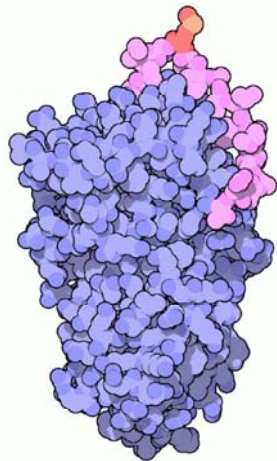
Alberta Health
Services

Promising Research

Omni Bio is currently evaluating Alpha-1 Antitrypsin to treat a variety of new indications

Omni Bio Pharmaceutical, Inc. ("Omni") recently announced that the Barbara Davis Center for Childhood Diabetes at the University of Colorado Denver has received IND regulatory clearance from the U.S. Food and Drug Administration (FDA) to initiate a Phase I/II clinical trial evaluating Alpha-1 Antitrypsin ("AAT") in Type 1 diabetics. Omni is sponsoring the clinical trial.

AAT, also known as augmentation therapy, is an FDA-approved, off-patent



Alpha-1 Antitrypsin

treatment currently indicated for the treatment of pulmonary emphysema among those with the alpha-1 antitrypsin deficiency. Preclinical studies based on OMNI's animal studies demonstrate that AAT may be effective in treating Type 1 diabetics and a variety of medical disorders.

Source: the complete press release and live audiocast and replay of the presentation are available on OMNI's website at www.omnibiopharma.com

Seizure drug could treat the liver scarring of Alpha-1

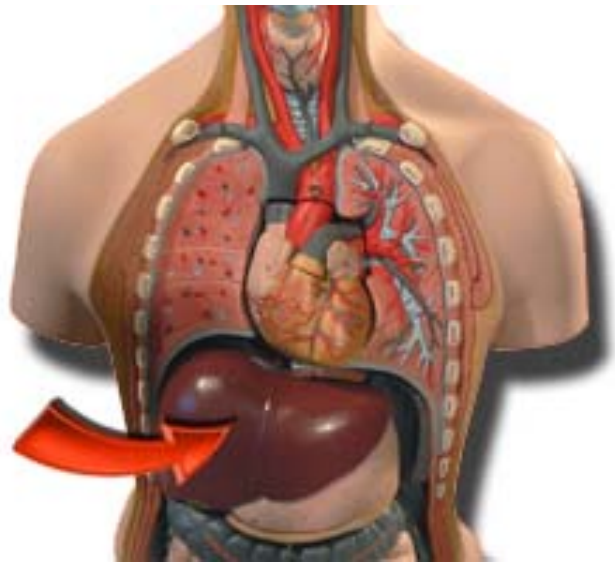
The Children's Hospital of Pittsburgh of UPMC and the University of Pittsburgh School of Medicine are exploring the possibility that the liver scarring of alpha-1 antitrypsin deficiency might be reversed or prevented with a medication that has long been used to treat seizures. This is particularly encouraging as alpha-1 antitrypsin deficiency is the most common genetic cause for which children undergo liver transplantation.

In the classic form of the deficiency, a gene mutation leads to abnormal protein. For the study on mice bred to have alpha-1 antitrypsin deficiency, the ZZ cell line was treated with carbamazepine, a drug used primarily to treat seizure disorders. According to Dr.

Perlmutter, from the Children's Hospital, "The most amazing finding was that the drug reversed the fibrosis in the livers of the mice and, after two weeks of treatment, the liver tissue resembled that of a healthy mouse." This is

particularly encouraging as alpha-1 antitrypsin deficiency is the most common genetic cause for which children undergo liver transplantation.

Source: www.sciencecentric.com



alpha-1 antitrypsin deficiency is the most common genetic cause for which children undergo liver transplants





January 2010:
Alpha and double lung transplant recipient, John Byrne carries the Olympic torch in Alberta



At 63, Alpha Syl Gerritsma wins bronze in the 100 metres with a time of 15.5 seconds at the Ontario Masters Track and Field competition

New gene technique can identify rare diseases quickly

A research team led by Dr Nada Jabado at the McGill University Health Centre research institute and Dr Jacek Majewski at McGill University has proven for the first time that it is possible to identify any genetic disease in record time thanks to a powerful and reliable exome sequencing method.

The exome, a small part of the genome (2%), is of crucial interest with regard to research on genetic diseases as it accounts for 85% of mutations.

“With this new approach, we no longer need to access patients who share the same altered gene pools to be able to identify the gene responsible for a disease. All we require are two persons affected by the disease not necessarily from the same family,” ex-

plains Dr Jabado, Associate Professor of Paediatrics at MUHC’s Montreal Children’s Hospital. “Now, within two weeks and with just two patients, we can easily isolate a gene. This compares to a time frame of six or seven months or even years before we saw results with the old process. This is really a positive breakthrough in genetic analysis.”

In their study, the researchers focused on isolating the mutation responsible for a rare and deadly genetic disease called Fowler Syndrome, where the brain fails to develop properly and usually dies before birth. Their results have revealed — between two patients with no family ties — a rare case of four mutations in the same gene. This illustrates well

the effectiveness of this sequencing technique, the goal of which is to isolate genetic alterations in cases of hereditary diseases among children, regardless of how prevalent they are in society.

“These results are very promising. There is now hope that in the near future we can treat a patient presenting a rare, unknown genetic disease in our laboratory, and within a few days be able to sequence his or her DNA to find the mutation that caused the disease,” states Dr. Jacek Majewski, Assistant Professor at McGill University’s Department of Human Genetics.

Source:
www.sciencecentric.com/resources/resource-000286-p-1.html

Alpha-1 Testing Reminder

Early detection of Alpha-1 is critically important. Once you know you have Alpha-1 you can make important lifestyle changes and receive treatment from your doctor to slow the progression of the disease.

The only way to detect Alpha-1 is through testing. We encourage you to take charge of your healthcare, take advantage of the new speedy, easy process and contact your physician with this critical new information. Whether you have been diagnosed or are awaiting testing please keep your physician informed. Print this page and bring it with you to your next doctor’s appointment.

And please, inform your close blood relatives that

testing and diagnosis is now quick, easy and free.

Once your doctor has determined that your AAT level is low (1.5 g/L or less, or below the normal mean for the testing laboratory) he or she can call the bilingual call-centre at 1-877-3 ALPHA1 (1-877-325-7421), to order a test kit. After your doctor receives the kit, three dry blood drop samples are taken then mailed to a testing facility at the University of Florida. Within two to three weeks the doctor will receive the results through the mail.

It is important to understand that while your personal information and test results will only be divulged to your doctor, unlike test kits that were available a

few years ago, these kits are not anonymous. Since the kits and results can only be sent to your doctor, he or she will place your results in your medical file (please see our web page on “Ethical Issues” to learn the implications of this fact.

Talecris, the makers of Prolastin® provides the specimen collection kit (including a postage paid mailer pre-addressed to the testing facility) and the testing services of the University of Florida Alpha1 Lab at no charge.

This program is managed and operated for Talecris by third parties; Talecris will not have access to anyone’s test results.

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Visit us on the web
at 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

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