



A Canadian PBC Society Newsletter

2010 - VOLUME 3

Toronto Area Meeting

On October 2, 20-25 members and family met to hear Dr. Gideon Hirschfield, Director of the Toronto Western Hospital's Liver Centre, aided by Catalina Coltescu, Research Coordinator, give an informative talk bringing us up-to-date on the genetic research into the causes of PBC. Dr. Hirschfield began by giving an historical overview of the disease, noting that currently researchers are investigating, to different degrees, three potential triggers: genetic (his own research area of interest), environmental and viral.

On the genetic front, following last year's breakthrough in isolating genes associated with PBC, further work has confirmed that certain of these genes have genetic overlap or involvement in other autoimmune diseases commonly found in patients with PBC or their families, including lupus, asthma, Crohn's disease, type 1 diabetes, sclerosis and Sjögren's syndrome. The evidence shows that there are several shared autoimmune susceptibility genetic loci.

On the treatment side, while ursodeoxycholic acid (urso) is a well-tolerated and effective drug for more than half of those treated with it, the search continues for new treatments (or treatments that are already on the market for a related autoimmune disease) that will be effective for those who do not respond well to urso, or ideally that will eliminate the disease. It is hoped that further clinical trials of the promising new drug INT-747 will take place next year.

After answering questions from members, Dr. Hirschfield and Ms. Coltescu were presented with gift certificates to thank them for giving up their Saturday morning for us. Thanks are also due to Mickie Nikoras for organizing the refreshments and Katherine Fuerst for organizing the room and coffee.





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The Canadian PBC Society - Ontario Region is Reaching for the Cure.

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Canadian PBC Society - Ontario Region Check out this interesting article on Night Blindness

Night blindness in primary biliary cirrhosis -- Waqar et al. 182 (11): 1212
-- Canadian Medical Asso
www.cmaj.ca

November 26 at 12:27pm · Share

PBC Society on Facebook

The Canadian PBC Society has now got its very own Ontario Region and Alberta North Region pages on Facebook. The pages are interactive tools for all of us – anyone can put up a tip, ask a question, recommend an interesting article or find out about events. Tara Kilmartin, who posted and maintains the Ontario Region page says ***"I believe the Facebook Page will be as good as the amount of action it gets; considering that the subject matter is quite serious, it is nice to have an online place to stay in touch, maybe meet new friends and develop the community. Be warned, though – it's great company and can be addictive. If you have not already looked at the page and clicked "LIKE" to follow our progress, please do so today".***

Thanks to Tara and Shauna Vander Well for giving us a presence on Facebook. If anyone would like to develop a Facebook page for their own region, please contact the regional representative.

Research

NIGHT BLINDNESS IN PRIMARY BILIARY CIRRHOsis

Mr Salman Waqar Mr Daniel Byles West of England Eye Unit, Royal Devon and Exeter NHS Foundation Trust, United Kingdom Night blindness (also called nyctalopia) is defined as poor eye adaptation to darkness. Typically patients might report difficulties with tasks at night or in dark places, such as trouble walking in dim lit rooms (e.g. movie theatres). Patients may also report difficulties driving in low light, at dusk, or in foggy conditions alongside a prolonged period of time needed to adapt from light to dark. The most common causes of nyctalopia include vitamin A deficiency and an inherited progressive retinal disorder called Retinitis Pigmentosa. Whilst retinitis pigmentosa has no association with Primary Biliary Cirrhosis (PBC), vitamin A deficiency can occur in up to 33% of patients with PBC. This is because PBC is a chronic liver disease characterized by ongoing inflammatory destruction of bile ducts eventually leading

to the development of biliary cirrhosis. Because of bile stasis in PBC, an inadequate quantity of bile salts is delivered to the intestines which results in vitamin A malabsorption and deficiency. In the body, vitamin A is present in its active form called retinol. This is essential for transmission of signals across the photoreceptor layer of the retina especially in low light conditions and thus its deficiency can lead to night blindness. Nyctalopia in patients known to have PBC should be investigated by checking serum vitamin A levels in the first instance. An examination by an optician and ophthalmologist should also be undertaken to rule out more common causes of difficulty seeing in dim lights e.g. refractive errors and cataracts. The ophthalmologist can carry out more specialized tests such as Electrotoretinograms (ERG) to confirm the diagnosis. During this, a corneal contact lens electrode is placed on the eye which is well tolerated by most patients. The retina is then stimulated by a brief flash of light that generates various traces which are recorded. Analysis of these can greatly aid with the

diagnosis. Once the diagnosis is established, simple measures such as regular vitamin A supplementation by the treating physician can reverse the nyctalopia in most cases. For more information on the role of ERG's please read our previously published report in the Canadian Medical Association Journal at <http://www.cmaj.ca/cgi/content/full/182/11/1212>

A pan-American study of the genetics of PBC STUDY UPDATE

Liver Clinic, Toronto Western Hospital, Toronto;
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Participation is voluntary and does not impact on any aspect of standard care. Important new evidence confirm that genes involved in Primary Biliary Cirrhosis (PBC) have involvement, in other autoimmune diseases commonly found in patients with PBC and their families.

How you can help...

Primary biliary cirrhosis (PBC) is a chronic, liver disease for which there is no cure and current treatment is far from satisfactory. The development of better treatment and preventive strategies in PBC is hindered by lack of understanding of the fundamental cause(s) of this disease. It is more common in family members than by chance alone suggesting important genetic contributors to disease. We are working hard to remedy this lack of information, and our study aims to identify the genes that lead to PBC and influence the outcome of this disease. Building upon a previous study in 2009 and published in the New England Journal of Medicine, our team conducted subsequent genetic tests on over 1,300 individuals with PBC and 1,800 non-PBC patients to identify potential risk loci, or genetic areas of disease susceptibility. Specifically, our findings show that genetic risk or "hot spots" for PBC are also hotspots for other autoimmune diseases such as systemic lupus erythematosus, systemic sclerosis, Sjögren's syndrome, asthma, and psoriasis.

It is imperative that we strengthen and extend these findings and, to do so, we need to undertake further scans of DNA sequences from many more patients. Our subjects are being recruited through collaboration with colleagues across North America and Europe. In Canada specimen collection has been generously funded by The Canadian PBC Society.

We have recruited over 1300 patients already but want to reach our target of 2000 cases.

We appreciate your help!

If you would like to consider taking part in this study please contact our Research Coordinator:
Catalina Coltescu at **416.603.5832** or by e-mail at catalina.coltescu@uhn.on.ca

INTERCEPT PLANS FOR ITS PHASE 3 PROGRAM IN PBC

Earlier this year, Intercept Pharmaceuticals, Inc. announced the results from an important clinical trial using a drug now called "obeticholic acid" or "OCA" (previously known as INT-747) to treat patients with PBC who are also being treated with UDCA. The combination of the two drugs showed a significantly greater improvement in disease markers compared to UDCA alone. These data were presented – on behalf of Dr. Andrew Mason, from the University of Alberta, and the other participating Investigators – at both of the major European (EASL) and American (AASLD) liver disease conferences this year.

Another study of OCA as a monotherapy has recently completed. Intercept hopes to find that OCA is effective as a single treatment in patients with PBC who are not taking UDCA. The results from this study are not yet available, but it is hoped they will be before the next EASL meeting in March.

Intercept is committed to supporting larger follow-on trials to prove the effects of OCA in PBC. The company has met with the U.S. Food and Drug Administration (FDA) and other regulatory authorities and is planning to start an international Phase 3 study by the second quarter of 2011.

Useful links:

<http://clinicaltrials.gov/ct2/show/NCT00570765?term=Primary+Biliary+Cirrhosis&rank=19> <http://www.pbcfoundation.org.uk/Home.htm>

<http://www.torontoliver.ca>

Our thanks to Cathi for keeping us informed.
Cathi I. Sciacca Sr. Director, Clinical Operations
Intercept Pharmaceuticals

Regional News

BRITISH COLUMBIA

This year, we had lunch meetings in the Spring and Fall for the Greater Vancouver/Lower Mainland Group. Both were held in New Westminster to accommodate members coming from all over the lower mainland. Ten of us got together at the Spring luncheon and four in the Fall. The Fall meeting stimulated some interesting discussion on the new research from Britain on PBC and muscle fatigue, and a lively discussion about the role of diet and supplements in PBC.

We are all anxious to hear about any new PBC research, and looking forward to our next get-together in spring (date and location to be announced). Any new members interested in joining us are asked to contact Kathryn Swift at kswift@pbc-society.ca or **604-988-4030**.

ALBERTA NORTH

Our 4th Annual Fall Barbecue was a big success. In attendance were our hosts, Garry and Saule, Garry's daughter, Cindy, Deb and her friend Gery, Gerald and Kathleen (all the way from Grande Prairie), and Shauna and Greg, and their daughter, Emily.

As always, it was an amazing feast, with enough left over to feed another army. We, of course, compared stories about symptoms, treatments and effective ways to manage. We ran a HIGHLY competitive silent auction which raised \$320 for research. Donation cheques are now winging their way to the National Office.

Thanks to all our donors: Shauna and Greg, Deb and her mother, Garry and Saule, Gerald and Kathleen, and Carol and Mike!! Contact Shauna Vander Well at AlbertaN@pbc-society.ca or **780-962-6217**.

MANITOBA

On November 12, four women met for lunch at the Red Lobster Restaurant in Winnipeg. We shared our stories while enjoying the good food (fabulous garlic cheese biscuits). One of us who has had a transplant told of her experiences, both the good and the difficulties of

the process. We discussed the fact that many of us have more than one auto-immune disease; I was diagnosed with MS over two years ago and others have Sjogren's syndrome, psoriasis and lupus. We welcomed a new friend from Portage La Prairie and she has a contact just across the border in North Dakota, so our numbers have increased to eleven. Now if we could all get together at the same time! We are planning a late Christmas gathering at my home on January 14.

I found an article on transplantation in the Sept./Oct. issue of The Wave, published by the Winnipeg Regional Health Authority, which may be of interest to the local group and perhaps all across the nation. Have a blessed Christmas with family and friends. Contact Carol Seburn at manitoba@pbc-society.ca or **204-254-5226**.

ATLANTIC

We had a great turnout for our Fall Business Meeting and luncheon with 16 attending. Information from the PBC Society and about our group members was given. We were pleased to welcome and give information and support to three new members.

We will contact the NS chapter of the CLF for information on their upcoming education sessions and encourage members to use their excellent programs for education and support purposes. We will continue to send information packages and have personal contact with new members as well as send cards to other members in need of support.

We will meet for a Spring social and use e-mail and the phone committee to keep up-to-date. Our last bingo bowling party was such a hoot that we are planning to do it again for our next June social. The gutter ball counter was the busiest of all of us, but at least we got some exercise and had a few laughs.

Wishing all a great Christmas and New Year. Take care till next time. Contact Judi Pemberton at atlantic@pbc-society.ca or **902-798-5554**

