CFA
CANADIAN FABRY ASSOCIATION

volume 7 issue 2
spring 2016

NEWSLETTER

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3RD FIN FABRY EXPERT MEETING

Gina Costantino

In November 2015 I had the pleasure of 'crossing the pond' to attend the 3rd Fabry International Network (FIN) Fabry Expert Meeting. The meeting was held about an hour north of London, EN UK at Latimer Place, Chesham, Buckinghamshire.

2015 marked the 10th anniversary of the FIN. The primary aim of the Fabry International Network is to facilitate collaboration between patient organizations around the world to support those affected by Fabry disease. The meeting, which ran from Thursday, November 19th through Sunday, November 21st, was the largest attended FIN expert meeting, bringing together over 45 individuals representing organizations from 5 different continents!

On Thursday evening, there was a group dinner where everyone had a chance to mix and mingle and introduce ourselves to one another. It was a wonderful opportunity to finally meet many of my Fabry 'friends' which I had gotten to know and collaborated with, while connecting through Facebook and the internet. Over the days that followed, I got to know representatives from Australia, Russia, Poland, Egypt, South Africa, Netherlands, Spain, Norway and Japan etc. including our friends Jerry & Jack from the USA. It was very exciting to meet and speak to so many heads of Fabry organizations from around the world – and even better that the majority of them spoke English very well :)

Friday's agenda was very full. The day started with a greeting and opening remarks from FIN's President,

CANADIAN FABRY NATIONAL PATIENT MEETING

Julia Alton
Vice President

It's that time again… the chance to stimulate our minds, share a room with those who understand the word "angiokeratoma," and the opportunity to take more control over our disease as we gain more knowledge, tools, and strategies to cope.

Join us at the Friday Night Social as we kick it off with live music. It's a chance to reminisce with fellow Canadians, a chance to build new connections, and a chance to welcome new patients and their families into the Fabry community.

Don't stay up too late as Saturday will be a jam-packed day, the lineup of speakers is exceptional bringing to us the latest updates in Canada. Don't forget your walking shoes as we will step out onto the Halifax Boardwalk together and enjoy "the Walk of Hope," as we walk for better days ahead.

The weekend will come to a close as we unwind with our families and friends over dinner.

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Christine Lavery. She went over the day's agenda and then introduced the first of many guest speakers.

Professor Atul Mehta
Dr. Mehta works at the Royal Free Hospital, in the Lysosomal Storage Disorder Unit, at the UCL School of Medicine in London England. His topic was "Changing patterns of natural history of Fabry disease". Dr. Mehta made note that Fabry is a progressive, multi-systemic condition affecting both males and females. In Fabry disease, the deficiency of the a-GalA enzyme has widespread effects throughout the body: on the brain, eyes, heart, lungs, kidneys, skin, GI tract and peripheral nerves. Dr. Mehta referred to GLA gene as a 'housekeeping' gene and that it is expressed in every cell in every tissue of our bodies. The Royal Free Hospital cohort is studying 59 different mutations falling into the following categories: Null, Classic (having < 1% enzyme levels), Missense and Late Onset mutations.

Dr. Alessandro Burlina
Dr. Burlina works at the University of Padova in North Eastern Italy. His topic was "Neurological Complication in patients with Fabry disease". He noted that there are many neurological complications associated with Fabry disease. Dr. Burlina spoke about different types of pain (evoked, attacks, crisis & permanent), cerebrovascular (cvs) disease (stroke), and neurovascular disease including cranial nerve involvement and excessive daytime sleepiness. He spoke about where Fabry patients typically feel pain but also noted that recent findings are showing commonalities in patients feeling pain in joints (knees & elbows) as well. Dr. Burlina noted that hearing loss in Fabry patients may be the bridge to cvs and that a sudden loss of hearing by Fabry patients should be thought of as 'bad' and brought up to your doctor immediately.

Dr. Gabor Lindhorst
Dr. Lindhorst is an internist-endocrinologist and a Principal Educator with the Academic Medical Centre in Amsterdam, Netherlands. His topic covered "Current management strategies in Fabry disease". Dr. Lindhorst spoke on what he believed to be the most current issues with the management of Fabry disease: Correct Diagnosis; Supportive Treatments and ERT. He noted, when it comes to Fabry disease, the medical community poorly understands: effective treatment of pain; when to start ERT; cognitive function including psychological / psychiatric diagnoses; ERT outcomes; and supportive care. He hopes to be able to one day perform a study using higher levels of enzymes, to lower antibody levels with immunosuppressants, to know the outcome of early treatment with ERT, and to develop a Fabry APP to capture day-to-day pain scores to improve pain management in Fabry patients.

Dr. Mehta spoke again – this time on "Indications for ERT in Fabry disease". The question of 'when should ERT be commenced?' was addressed in his presentation. Dr. Mehta noted the following: 1. Be certain of diagnosis 2. Males with 'classic' Fabry disease should be started as soon as possible 3. Females with intractable symptoms or first sign of organ damage 4. Children – rarely need to treat girls/non-classic boys; classic boys – once symptomatic. There was a brief discussion among many of the medical personnel in attendance with good consensus that Enzyme Replacement Therapy should start before there is measurable end-organ damage in 'classic' Fabry disease patients.

Dr. Gabor Lindhorst spoke again – his next topic was "Stop criteria ERT incorporating cardiac progression" which is a real scenario in the Netherlands. He described cardiac disease progression in Fabry patients as problems with function (heart failure), valves (leakage), rhythm and blood flow (coronary). Dr. Lindhorst spoke about options of treating rhythm abnormalities with anti-coagulants, pacemakers and implantable cardioverter defibrillator (ICDs) instead of ERT. He spoke about valvular replacement surgery; more vigorous follow ups with Echos and MRIs, EKGs & Holter analysis, and better use of ACE inhibitors, anticoagulants and diuretics.

Dr. Uma Ramaswami
Dr. Ramaswami is a Metabolic Pediatrician, in the LSD Unit at the Royal Free Hospital in London England. Her discussion was titled "Recognizing the early signs and symptoms of Fabry disease and unmet needs". The doctor noted that 70% of children will manifest acroparesthesia in the 1st decade of life. Gastrointestinal manifestations are among the earliest of symptoms for Fabry disease. Mutations resulting in loss of function with no residual enzyme activity correlates with early onset retinal vessel tortuosity and early onset tinnitus correlates with disease severity in children.

Professor Christian Hendriksz
Dr. Hendriksz is currently a Professor of Paediatrics and Child Health at the University of Pretoria, Manchester UK and a Consultant in Transitional Metabolic Medicine in the Metabolic Unit of Salford Foundation Trust (Hospital and Health Care) in the UK. His topic was "Therapeutic goals in the treatment of Fabry disease". His particular interest is in aiding children in transitioning from paediatric care of Fabry disease to adult care. He focuses greatly on the therapeutic role of treating the disease and not the diagnostic specialty. For example, a therapeutic goal for treating kidney disease may be a person's desire not to go on dialysis, while a physician's diagnostic (treatment) goal would be to reduce GFR to within normal range. Both 'goals' may be achieved using similar therapies but outcomes appear different. Dr. Hendriksz believes the need of adult patients affected by rare inherited metabolic disorders is even greater than for children and that there is a lack of knowledge in this area. His goal is to make a significant difference to the lives of children and adults affected by rare inherited metabolic disorders.

After a wonderful lunch break where we were given time to discuss further the morning's session, we returned to the conference room to hear updates from representatives of Amicus Therapeutics, Genzyme (a Sanofi Company) and Shire Pharmaceuticals. The updates were followed by a question and answer period and then the Board of Directors of FIN held their AGM. Attendees were given a couple of hours to themselves to rest or roam the grounds. Everyone reconvened for the 10th Anniversary Celebration Dinner before retiring for the night.

Saturday's meeting began after breakfast with Jack Johnson, the VP of FIN and Founder / Executive Director of FSIG in the USA greeting everyone, outlining the half-day agenda and the list of guest speakers for the morning.

The first two speakers were from the UK MPS Society and discussed a recent Pedigree study result, as well as best practice pedigree analysis.

Dr. Uma Ramaswami spoke again, this time on the topic of "When to test asymptomatic children and when to start treatment". Dr. Ramaswami reported that The European Fabry Working Group consensus document Biegstraaten et al 2015 stated "Pediatricians in the group in particular concurred in their view that early initiation of ERT in asymptomatic children in the context of a very slowly progressive disease is likely to interfere with normal childhood". The conclusions were that onset of symptoms in Fabry disease are early but slowly progressive and there is a point of reversibility of symptoms so monitoring children is the key message.

After another question and answer period, presentations were given by a few Patient Organization Group members.

The first was by Anna Meriluoto, a Board of Director of FIN, VP of Finnish Fabry Society and VP of Rare Disease Organization HARSO. Anna spoke of the Global Perspective of Fabry patient organizations and identified the 5 A's of each organisation: Advocacy, Awareness, Action, Access and Authorization. She also acknowledged some common challenges facing all patient organizations: volunteers as the work force, lack of good volunteers, geography and the disease itself.

Next up was Lea Chant from Fabry Australia who spoke about the special challenges for Fabry patients living in Australia. Their treatment options are currently Fabrazyme and Replagal. Amicus' Migalastat HCl Chaperone is being evaluated for approval. In Australia, children are not automatically treated, there is little cohesive coordination of patient care across the country, there is no national rare disease plan although "Rare Voices Australia" is gaining traction, and funding for treatment drugs is constantly under budget pressure. A children's book called "Faber: a dragon" was written and published by author Julie Fitzgerald in Australia as a simple explanation for her niece's friends to understand and help her with her Fabry disease.

Then Mary Pavlou spoke about the "Daily life challenges for Fabry patients in Greece". Mary is a Registered Nurse and the Secretary of the Hellenic Association for Lysosomal Diseases which has 142 members consisting of patients with Gaucher, Pompe, MPS and including 27 with Fabry disease. Among many common challenges, in Greece they do not have 'reference centres' or specific hospitals where specialists for these diseases can be found. While ERT is available by prescription, and patients must travel sometimes very long distances to fill their own prescriptions, home infusions are illegal. Patients must

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bear an additional charge for use of outpatient services and hospital services.

Lastly, Mr. Jun Okada gave a brief presentation about the Japan Fabry Association being established in 2002. It has 435 members, representing about 120 families. Their philosophy is to achieve safe and effective treatment, reliable systems and an inclusive, accepting society. The JFA hold seminars and symposia in 6 main districts of Japan. They currently have Fabrazyme and Replagal for ERT treatments and are involved in clinical trials with Chaperone Therapy (Amicus) and Biosimilar ERT (JCR). In 2015 a new law was enacted for intractable and rare diseases which: designates more diseases as intractable and rare, provides budgeting for subsidy of medical expenses, guarantees the livelihood of patients, safeguards patients' rights, and transfers authority from central to local governments. Jun then gave a brief outline of what has been planned for the next FIN Fabry Expert Meeting and expressed his wish for all to attend in Tokyo, Japan in May 2016.

Christine Lavery gave closing remarks, thanking all for their attendance and thanking especially all those that came from very far away. She invited everyone to take some time to visit the Expression of Hope art exhibition in the next room. The exhibition featured contributions of artwork supplied by people affected by LSDs around the world and many pieces were truly remarkable and very moving.

This concluded the Fabry International Network Fabry Expert Meeting This meeting and all discussions provided great insight into how Fabry is dealt with in different parts of the globe; what therapies are being utilized, what research is being done, how patients access health care and ERT differently depending on where they live etc. It was quite the distance to travel and a very demanding agenda over just 2 ½ days, but very well worth it when considering the networking opportunities gained and the information gathered and shared with all.

THANKS TO SUPPORTERS

We would like to thank all of our supporters that helped make this newsletter possible.

We have received financial support from the three drug companies that are currently providing hope for Fabry patients through their research and the products they provide.

Amicus Therapeutics

Shire

genzyme

We would also like to thank all of the physicians, specialists and medical professionals that have helped in so many ways. From providing guidance on medical terms and details to caring for members of our community every day.

And of course we would like to thank all of the patients and family members that have volunteered their time and energy to assist in all the many ways that are necessary in the creation of such a large effort. It is through their efforts that we hope to inform and build a community of Fabry patients for the benefit of patients, their families and caregivers.

WEBSITE UNDER CONSTRUCTION

The Website Committee
-Gina, Ryan, Donna, Julia and Taimi

Hello Everyone, We are working hard with some professionals on a redesign of our website. This site will feature dynamic content that can be easily updated to keep everyone as up to date as possible. This site will make it easier than ever to manage your CFA membership and register for our meetings.

This website will conform to the wide variety of devices used to connect to the internet, be it a phone, tablet, desktop or laptop. This means less scrolling and faster access to the information you want to see. We are looking forward to the launch and hope you are too.

REMEMBERING FRIENDS

The CFA is considering including remembrance notices for Fabry Patients and Accociation Members that we have lost over the years. To give us information on those to include, contact us at: secretary@fabrycanada.com

BECOME A MEMBER

Would you like to become a member of the Canadian Fabry Association? It will ensure you receive the most current information concerning your disease and its treatment. All information will remain completely confidential and will be shared with no other organization.

Date: _____ 20 _____

Name: _____

Address: _____

City: _____

Province: _____ Postal Code: _____

Home Phone: _____

Work Phone: _____

Email address: _____

Individual amount $15.00
or Family membership (family living in the same home) amount: $25.00

Complete form and mail to:

Canadian Fabry Association
52 Glen Forest Drive
Hamilton ON
L8K 5V8
or register online by visiting our website:
www.fabrycanada.ca

MAKE A DONATION

Would you or a family member like to make a donation so that we can continue to educate and advocate for the best treatment as well as communicating with and for Fabry patients in Canada?

The Canadian Fabry Association (CFA) is a registered not-for-profit organization. Recently, through the generosity of one of our Board Members, the registered charity The Fabry's Charity Association has become available for the CFA's use. If you are interested in making a charitable donation and would like a tax receipt, please make you cheque payable to The Fabry's Charity Association.

100% of donations to the CFA are used to promote education, patient support and access to treatment for Canadian Fabry patients. You can make donation cheques payable to The Fabry's Charity Association and mail the cheque to us.

Send the cheque to:
The Fabry's Charity Association
52 Glen Forest Drive
Hamilton ON
L8K 5V8
or register online by visiting our website:
www.fabrycanada.ca

Thanks for your donation to the CFA! It goes to help Canada Fabry patients, their families and caregivers.

Live Well with Fabry