canadianfabryassociation
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newsletter

VICTORY JUNCTION
Fabry Family Weekend

Julia Strauss

A weekend of meeting other families like the one I grew up in…. a weekend of leaving behind worries and fears and living in the now… a weekend of seeing Fabry patients in the spotlight and on the dance floor… and a weekend to remember the ones we've lost to Fabry.

In September I volunteered at a camp called Victory Junction in North Carolina, it was a weekend where Fabry patients came together as a family and took a break from the struggles every day life brings them. The camp had every sport, and every activity one could wish for!! Bowling, fishing, arts & crafts, archery, baseball, the body shop (go and work on real NASCAR vehicles), and even a hair salon!

To look around me and see the athleticism, talent, and skills of so many other patients was a great feeling to experience! Seeing a patient up on stage playing guitar and singing to all of us made me proud and thankful that I had the opportunity to meet so many incredible individuals. What amazed me the most however is that it was only a decade ago that I first met a Fabry patient other then my Father, and when my Father was growing up, he didn't know one other person with the condition he had. And there I was sitting in a room with 40 – 50 other Fabry patients of all ages and I realized that even though I live with a rare condition, I am fortunate enough to grow up knowing others who face some of the same struggles and challenges that I have, and fortunate to grow and learn with these people as well.

I hope that one day we will be able to offer a weekend getaway for Canadian families and give them the same incredible experience!

The 2012 National Patient Conference

Gina Costantino

The 3rd Canadian Fabry Association (CFA) National Patient Conference was held June 8/9 2012 at the Westin Nova Scotian in Halifax, Nova Scotia. The "Live Well with Fabry" Conference was a tremendous success.

The CFA hosted a Welcome Reception and an evening social prior to the Conference, where many patients, family, friends, doctors, presenters and Sponsor representatives had a chance to meet and mingle, chat and socialize. The following day was full of interesting speakers, who provided important government and medical updates, strategies for diet and nutrition, and emotional coping skills and support. We incorporated a few new things into the programme as well. Live French translation was provided for all the presenter's speeches, as well as all conference documentation. Daycare was provided, which the little ones seemed to enjoy. A Youth Forum was held and those in attendance found it invaluable. Connections were made and new friendships were formed. The "Walk for Hope" saw virtually all in attendance out for a walk along the beautiful Halifax boardwalk. The walk provided us all with some fresh air, beautiful scenery, additional opportunities to socialize and ultimately, a wonderful sense of community.

Our Keynote Presentations began with Diane McArthur, the Assistant Deputy Minister and Executive Officer of Ontario Public Drug Programs. Ms. McArthur addressed current and future access to Enzyme Replacement Therapy (ERT) in Canada. She provided an overview of the Canadian Fabry Disease Initiative (CFDI), efforts made to date by the provinces and territories to support drug access for Fabry patients across Canada, and more importantly, beyond the September 30 2012 expiration of the CFDI, what future access to ERT would be. Ms. McArthur noted that discussions had been well under way with drug manufacturers and the CFDI consortium to expand/continue the study and to continue collecting data. She was happy to announce that while all the 'next steps' were being figured out, there would be no interruption for patients receiving ERT.

Dr. Sandra Sirrs, the Medical Director of the Adult Metabolic Diseases Clinic at Vancouver General Hospital and the regional site investigator for the CFDI in British Columbia gave the next presentation. Dr. Sirrs spoke about the 5 years of data collection and Canadian criteria needing to evolve as new data emerges. She addressed issues that were beyond the control of the CFDI and why there had been so many amendments to the study (Genzyme shortages, Shire's new manufacturing process, and changes to Canadian laws). Within the control of the CFDI changes were also made – altered (reduced) frequency of tests, and the use of new tools (biomarkers). Dr. Sirrs reiterated the benefits of enrolment in the CFDI which included protection from drug shortages, protection from regulatory issues such as shipping and infusion, support for home infusions, centralized payment (no dispensing fee, no deductibles) and the surveillance of Cohort 1c (natural history patients, i.e. currently "well" patients). She was both extremely informative and an excellent speaker, even offering a bit of comic relief now and then. From post-conference surveys, 98% were very satisfied with her presentation.

Pamela Dill, a renal dietitian provided dietary strategies to aid in delaying the progression of kidney disease. Also included was a discussion of the nutritional aspects of heart health and a review of what adequate nutrition is required while on renal replacement therapy. As many Fabry patients have kidney involvement and/or heart involvement, Pamela's presentation was very informative to many. She also allowed time during the lunch break to have people do BMI testing on the scales she provided.

After the lunch break, Rick Sgroi, the current Vice-President of the CFA and an executive Board Member since the CFA's inception, spoke briefly about his own personal dealings with Fabry disease, including his recent kidney transplant. He presented Dr. Joe T. Clarke, a doctor who has spent his career investigating and helping Fabry patients and their families, a retirement gift on behalf of the CFA. While Dr. Clarke has retired from his practice, he continues to be the primary regional investigator for the CFDI in Ontario.

Dr. Gordon Butler, a clinical psychologist, gave a presentation on living well with chronic illness. He reviewed concerns and types of stressors that are part and parcel of dealing with Fabry, and discussed the impacts of these stressors on one's health. Dr. Butler provided coping strategies and ways to challenge negative thoughts and cognition. Some strategies included staying informed (to reduce anxiety); social networking (for emotional support); incorporating healthy behaviours (good nutrition and physical activity); cognitive therapy (scheduling 'worry time'); and any form of relaxation (deep breathing for some, vigorous activity for others).

The next presentation focused on updates on the development of Gene Therapy for Fabry Disease and was given by Dr. Jeffrey Medin. Among other credentials, he is the Principle Investigator of the recently awarded funding from the Canadian Institute of Health Research (CIHR) Emerging Team Project in Rare Diseases. In Dr. Medin's words "gene therapy has the capacity to correct disease" and if we can get enzyme into the genes and into the blood stem cells, then we can get the benefit to the whole blood system and body. He believes Fabry will respond well to gene therapy because it is a single gene disease as compared to other lysosomal storage disorders (LSDs). Dr. Medin noted the premise behind gene therapy is to have lower levels of enzyme continuously released in the body by way of metabolic cooperativity vs. receiving a single large bolus (ERT). The idea is the body cooperatively distributes enzyme throughout itself without disrupting any other of its systems. Dr. Medin reported that the Government is very enthusiastic and he is hoping clinical studies will begin within the next 2 years. He said there are three things we can do: be patient, be supportive and believe!

Mr. Jerry Walter was the final presenter. He is the founder, president and only staff member of the National Fabry Disease Foundation (NFDF), which is one of two patient-centered groups running in the United States. He spoke about the benefits of becoming and staying connected to the Fabry community including being able to achieve many more positive changes for individuals, families and the overall Fabry community by learning from each other and by working together. Jerry works very hard at providing support and assistance as well as helping to educate and raise awareness of Fabry Disease. He offered to take pictures at the conclusion of the Conference, of anyone who was interested in being one of his 'Faces of Fabry' – which is on the NFDF website. There was quite the line-up of people wanting to be one of Jerry's faces!

CFA Board President Darren Bidulka thanked all our presenters and all the medical personnel who had attended the CFDI Conference earlier in the week and chose to stay on and attend our Conference to give their support and personal time in dedication to our cause. Darren also thanked our Sponsors for their educational grants – Amicus Therapeutics Inc., Genzyme Canada Inc., Shire Human Genetic Therapies (Canada) Inc. and Bayshore Specialty Rx – without their support, the Conference would not have been possible. He noted we just may have had the largest gathering of Fabry patients, family and friends ever, which is a testament to the hard work of the CFA organizing committees and our star event coordinator, Sherry Sim, Innovative Business Solutions. But it also speaks to the value we all place on being together to support one another and come together as a community. If you weren't able to attend, we hope you can join us for future regional and national conferences. Everyone's participation made for a great weekend for all and provided a better understanding that one really can Live Will with Fabry.

Research for the 'Rare' Community

Darren Bidulka

Everyone in the Fabry community is well aware of the clinical research trials in which we are participating. Most every Fabry patient is enrolled in the Canadian Fabry Disease Initiative ("CFDI") and we receive regular updates on the CFDI at our patient meetings. I would say our Fabry community has a good understanding of the currently available treatments and the importance of research to our health.

But, what about other research into new and hopefully better treatments for Fabry patients? And, what about research for our bigger community of patient's with other rare disorders? We are members of a big community of patients with rare disorders that face many similar challenges – research focussed on a particular rare disorder can have benefits for many in the rare disorder community.

We are introducing a new series for our newsletters – Rare Disorder Research Updates. We plan to include an article in each newsletter highlighting a research project that affects the rare disorder community. Some articles will focus directly on Fabry while others will be on other rare disorders.

Research is so very important to our lives. Without research we would not have existing treatments available to us. Many in the Fabry community remember the days before enzyme replacement therapy existed. It is the combination of industry being willing to invest, government providing conducive policy, medical community support and patient's willing to participate in research clinical trials that creates advances in treatment. Without all of the ingredients progress is held back.

The Canadian Rare community has had two important announcements from government in the past year that will accelerate research. In February, the Canadian Institutes for Health Research ("CIHR") announced funding for nine research projects – all of them focussed on the rare disorder community. Then in October the federal government announced two initiatives – new Health Canada policies for authorizing treatments for rare disorders and second, the launch of the 'Orphanet' database.

The nine research projects announced by CIHR include one near to us – Dr. Jeff Medin is leading the research on gene therapy for Fabry patients. The other eight are a mix of research focused on specific disorders and others focused on issues common to many rare disorders. You can learn more at CIHR's website http://cihr-irsc.gc.ca/e/44945.html.

The October announcement on new Health Canada policies is a big win for the rare disorder community. We are members of the Canadian Organization for Rare Disorders ("CORD"). CORD has spearheaded education aimed at making our federal members of parliament aware of the need for change at Health Canada in order to deal with the many unique challenges faced by the rare disorder community. From the start Health Canada understood the unique challenges we face but it took the will of government to initiate the changes. We have worked for years to achieve this big step in the right direction.

Many of us have dealt with the pain and frustration of misdiagnosis. Unfortunately, the general medical community just has so many issues to deal with that it cannot know about every disorder. It takes rare disorder experts in the medical field to provide the care we need as Fabry patients. Imagine if every doctor had access to a website where they could enter symptoms and get suggestions of possible rare disorders? Well, the introduction of Orphanet is another big step forward as it does just that. Orphanet was pioneered in Europe and is now coming to Canada. You can take a look at http://www.orpha.net/national/CA-EN/index/homepage/ for the Canadian entry point or http://www.orpha.net for the European site.

These are exciting days with significant advancements and funds being dedicated to helping improve the lives of patients with rare disorders. We wish to the researchers' success, thank the medical community for their support of research and encourage patients with rare disorders to continue their individual support of clinical trials.
Take care, Darren.

My Fabry Journey

Christine Francey

Burning hands and feet, bad stomach pains, high fevers for no reason, these are a few of the symptoms my siblings and I thought were normal, because we all had them.

Rheumatoid Arthritis, Carpal Tunnel, Acid Reflux, Irritable Bowel, is only a short list of the many things that doctors told me I was suffering from, even though the tests were always inconclusive. The hardest to hear the doctors say, it's all in your head, you have anxiety, you are using excuses not to go to school.

Denial, anger, sadness, and then finally , proof that you really were sick all this time, are the things I felt when 3 years ago, Fabry Disease was finally diagnosed in my family!

So far there are 13 of us with a positive diagnosis, my Mom, myself, all 4 of my siblings and some nieces and nephews. The biggest heartbreak of all was that I had passed this on to my son. Colin was 10 when we found out.

I really didn't know what to do, and as I usually do when I don't know the answer, I looked it up on the internet. If you know all about it you can fix it, right? Well in our case, not quite. In the process of my search I met so many people who have been through the same thing and have helped me get through to a place where I have the strength to fight, to accept and to learn to live a completely different life.

The doctors told us there was no cure, but a treatment that would help us live better, healthier, longer lives. They also told us, that we were the first family in Manitoba with a diagnosis of Fabry Disease. This meant we were starting from the beginning to apply for funding for the treatment. Over 2 years, we saw many specialists, went for many tests, asked a million questions, and constantly wondered if the treatment would ever come.

In the meantime, my husband and I, and our 2 boys did everything we could to learn all about Fabry Disease. This search is what brought us to the Canadian Fabry Association, and the ever helpful, Darren Bidulka. We travelled to Calgary to the patient meeting in September 2011, and then to Halifax in June 2012, with the hope of finding some information and meeting other people with Fabry Disease.

When we attended our first family meeting in Calgary, we felt overwhelmed, not only with information but with the fact that were many others who lived with this disease. It was very hard for us to understand how all these people could be happy and going on with their lives like they were "normal". We just weren't there yet. We were sad, angry and afraid.

Our trip to Halifax in June was what really brought us hope for the future. We met and had the chance to ask questions of great doctors and nurses, who then helped us, get what we needed to finally begin treatment at the end of June. Another highlight in Halifax was meeting Jerry Walter and being so lucky to be invited to Victory Junction for the Fabry weekend.

Although being on treatment wasn't exactly what everyone hopes, there have been a few bumps in the road, it gives us hope. Hope that there will be improvements in my health, hope that Colin will never suffer the way others have without treatment and a great happiness that we have met many wonderful people. People who have been there for us and without them we would not be where we are today.

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

Bayshore Home Health

genzyme

Shire

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.

Quebec Patient Meeting

François Chabot

The Quebec patient meeting was held October 27th, 2012 at the Sacre-Cœur Hospital. A big thank you to Carole Fortier and to Claudia Menard of Dr. Bichet's office for all their help in contacting patients and booking the room. We had 16 patients, family members and health care professionals in attendance. Thank you to Nancy Gagnon (Shire) for helping us find a guest speaker and for taking time to support this meeting. Thank you to Denis Lacoste (Genzyme) for taking the time to meet me prior to the meeting.

Our guest speakers were Dr. Daniel Bichet (MD), who treats the QC patients, Marie LeFrancois, a nutritionist from the McGill Health Center and myself, treasurer of the Canadian Fabry Association (CFA).

Dr. Bichet provided an update on the results of the studies he has been conducting in the last three years. He spoke of the implications of the switch from Fabrazyme to Replagal and the switch back to Fabrazyme. He also informed us on the status of the CFDI study, as the previous agreement came to an end in september 2012.

The Amicus trials were also discussed by Dr. Bichet as it might be a good avenue for some patients with Fabry. Dr. Bichet also handled a question and answer period and had patients talk about their experience. I found this being the most pleasant part of his presentation as we got to hear from everyone that was attending.

Marie LeFrancois provided the patients with an interactive discussion which enabled us to have many answers to questions we had been having for a while. She also provided us with links to websites and literature to keep us informed to make wise health related decisions. We had been looking for a nutritionist for many years and we were delighted to have Mrs. LeFrancois come to our meeting and enlighten us on our food (and exercise) habits.

As for myself, I was happy to present some of the advances in Dr. Medin's research (University of Toronto) which is going forward because of a CIHR grant he received this year. We are all happy that tests on humans are imminent and we are eager to get feedback from this first gene therapy treatment for Fabry in the world! I also spoke about the future of funding for ERT in Canada as the CFDI was renewed for one year until September 2013.

The RQMO is also planning a web-conference in the near future to discuss psychological aspects of Fabry on patients. This subject was the most sought after in all of the comments sheet post conference. Stress, the feeling of being alone and guilt are some of the subjects that will be discussed during this web-conference.

Finally, thank you to the patients and family members that attended the meeting. It is always nice to meet and learn and share our experiences with others. We look forward to future patients meetings and hope more and more people will feel the desire to come and see that these meetings are very fulfilling.

Francois Chabot,
Treasurer of the Canadian Fabry Association.

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 _____

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

Complete form and mail to:
Canadian Fabry Association
PO 40036
4250, 1re avenue
Québec (Québec)
G1H 7H6
or register online by visiting our website: www.fabrycanada.com

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 registered as a not for profit organization but it is not a registered charity. To become a registered charity is quite expensive. Since the CFA is not a registered charity it cannot issue tax deductible receipts.

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 Canadian Fabry Association and mail the cheque to us or donate electronically online on our website.

Send the check to:
Canadian Fabry Association
PO 40036
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or visit our website: www.fabrycanada.com

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

For better days.