Soft Bones Canada

Newsletter

INAUGURAL ISSUE

A LETTER FROM THE PRESIDENT

In 1998, after years of typing in the now all too familiar word "Hypophosphatasia (HPP)", a hit finally came up on the internet! My daughter had been diagnosed in 1994 with this ultra-rare disorder (I would be diagnosed a few years later), and we were informed by physicians at that time that it was believed she was the only person alive with HPP. We were desperate for answers. Was she going to die? Would she have a normal life? Would she be in a wheelchair or require multiple surgeries? That connection with a fellow HPP family would ignite a passionate drive to ensure that other families would not have to navigate this difficult road of managing a rare disease with little support and few answers.

My name is Debbie Taillefer, and I am the founder and president of Soft Bones Canada. In 1999 out of our living rooms at home, Margery Callen and I formed the first Canadian organization called the "Canadian Hypophosphatasia Contact". This would begin a process of reaching out across the globe coming in contact with many wonderful patients throughout the world, not the least of which would prove to be a very important relationship for us, founder and president of Soft Bones, Inc., Deb Sittig from New Jersey, USA.

During this time the health of Margery, my faithful support and manager, began to decline. (CONTINUED ON PAGE 2)

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ANNUAL DATES

October 25-31
HPP Awareness Week

October 30
HPP Awareness Day

Last Day of February
Rare Disease Day

HPP

Hypophosphatasia (HPP) is a genetic, chronic and progressive ultra-rare metabolic disease characterized by abnormal bone mineralization that can lead to fractures and deformity of bones, profound muscle weakness, seizures, respiratory failure and premature death.

She had overseen the International Yahoo group and our website while my family had moved to the US and now she needed me to once again, take the helm. Back at home in Canada, I designed a new website and utilizing the acronym HPP, we became HPP Canada. I now prepared to moderate everything solo for the first time in 14 years. We determined that with the major shift to social media, it was time to retire our Yahoo mailing list. At the close of its usefulness in October of 2013, membership for the group had grown from 2 to 295.

Continuing with the strong connection and relationship we had formed with Soft Bones US, in 2013 Deb Sittig invited me to become Soft Bones Canada and come under the Soft Bones family umbrella.

Now, in 2015, we are very excited to announce that Soft Bones Canada has come into her own and with the support and investment of Soft Bones Inc., has begun yet another new chapter. We formally organized as a non-profit organization with our very first Board of Directors including Jennifer Boin, Director of Education and Events and Lisa McGuffin, Director of Patient Care Services. Joining with us is Dr. Cheryl Rockman-Greenberg, MD, CM, FRCPC, FCCMG Professor, Department of Pediatrics and Child Health and Programme in Genetics and Metabolism College of Medicine, Faculty of Health Sciences, who will be Chair of our Scientific Advisory Board.

This has been a banner year for HPP around the world but especially for Canada. Along with our maturing to have a full-fledged patient support group, we have our first approved therapy developed by Alexion Pharmaceuticals known as Asfotase Alfa, discovered by our very own Dr. Philippe Crine, Ph.D., Vice-President & Chief Scientific Officer, Professor of Biochemistry at the University of Montreal Medical School and a leading scientist in the field of molecular enzymology and protein engineering.

This has truly been an exciting year of firsts! However, we know there is still much work to be done and so our commitment remains as passionate and steadfast as it was almost 17 years ago: to provide valuable information, education and support to patients, their families and caregivers living with HPP. We hope you will join with us in promoting research, proper diagnosis, and access to therapies through raising awareness in the coming years.

Debbie Taillefer
PRESIDENT SOFT BONES CANADA

Dr. José Luis Millán, Deb Sittig and Denise Goodbar at the ASBMR conference in Seattle October, 2015.

Thank you to our friends and colleagues at Soft Bones (U.S.). Your support of our efforts to establish a non-profit patient organization in Canada has been incredible and much needed. We look forward to working with you and sharing the journey.

INTRODUCING SOFT BONES CANADA'S DIRECTORS

Jennifer Boin (Ontario)

DIRECTOR OF EDUCATION AND EVENTS

My name is Jennifer Boin and I am thrilled to be a part of this new and exciting undertaking. I was diagnosed with HPP at three years of age, but at that time no one seemed to know what to do with me. Luckily, aside from my dental issues, my HPP remained quiet for most of my childhood, but ramped up in my mid-twenties and continues to challenge me physically and emotionally.

Like so many other patients with HPP, I felt very isolated and frustrated by the lack of reliable information available to me. In addition, I had no one to support me, or offer advice about how to best care for myself. It felt like I was trapped inside this shell that was wearing down far faster than my spirit.

Luckily, I found Debbie and Lisa and together we hope to save other patients and caregivers from feeling alone in their search for information and a voice.

Aside from my work with Soft Bones Canada, I am a full time high school teacher, currently working as a guidance counsellor. I am the mother of two very active young children and the wife of an equally active and very supportive husband. I love to hike and read, but not at the same time – that's dangerous.

I look forward to working with all of you.

Lisa McGuffin (Manitoba)

DIRECTOR OF PATIENT CARE SERVICES

Hello, my name is Lisa McGuffin. I am one of three children diagnosed with HPP in our family. While my two siblings were affected severely as children, I have only begun to notice a great change in my thirties. Because I was determined to prove that this disease would not define or limit me, I completed a degree in nursing at UWO in London, ON, and currently work in long-term care.

My husband and I have seven (yes, seven) children, who are filled with creativity and fun! When we are not busy with our many jobs, you will find us playing the piano, gardening haphazardly, or reading.

Our family has dreamed of a treatment or cure for HPP for over 50 years, and it has been so exciting to watch the progress and possibilities of treatment break wide open here in Canada!

I look forward to being a part of the Soft Bones Canada community with you.

OUR COMMITMENT

SBC is a source of education, information, encouragement and support for Canadian individuals and their families affected by HPP, including interested individuals in the medical community.

- The first report of Hypophosphatasia (HPP) in the English-language medical literature has been attributed to Dr. Bruce Chown of the University of Manitoba, Winnipeg. In 1936, Chown described two sisters of Welsh descent with what he called “renal rickets” (Chown 1936; Fraser 1957).
- A Canadian physician, John Campbell Rathbun, first named the disorder Hypophosphatasia in 1948, after he treated a baby boy with paradoxically low levels of alkaline phosphatase. The boy was first treated on December 19, 1946. For a long time it was known as “Rathbun’s Syndrome.”
- In 1995, Margery Callen of Toronto, Canada, diagnosed in 1992 after 5 years of fractures and surgeries, connects to the internet seeking support. She discovers and makes first contact between patients Del Sieber and Carol Clapshaw in the US and they start communicating.
- In 1998, Margery and Debbie Taillefer connect. In 1999, they begin to discuss the need to provide support to Canadians and their families. They form the first Canadian organization called the “Canadian Hypophosphatasia Contact.” They develop the first Canadian website and brochure with a brief synopsis of HPP and support contacts.
- Dec. 23, 1999, Debbie creates the first international support group via Yahoo Groups called “Worldwide Hypophosphatasia Support Mailing List.”
- 2006 Prof. Philippe Crine, of Enobia in Montreal, PQ, developed the first therapy called “Asfotase Alfa” - a bone targeted enzyme replacement therapy for Hypophosphatasia.
- 2008 Dr. Cheryl Rockman-Greenberg, MD, CM, FRCPC, FCCMG Professor, Department of Pediatrics and Child Health and Programme in Genetics and Metabolism College of Medicine, Faculty of Health Sciences started the first clinical trials. Enobia flew a little girl from Belfast, Northern Ireland, to Winnipeg for treatment.

SCIENTIFIC ADVISORY BOARD

In order to ensure that we are providing the most accurate information about Hypophosphatasia, a Scientific Advisory Board has been established. This board currently includes members from a variety of medical, research and patient care backgrounds.

HISTORY OF ASFOTASE ALFA IN CANADA

This is my first official message to the Soft Bones Canada community. Eight years ago I received a phone call from Enobia Pharma, the Montreal-based biopharmaceutical company, asking if I were interested in participating in clinical trials of an innovative product, ENB-0040, that had been developed and had already shown great promise in pre-clinical trial studies on the treatment of the mouse model of Hypophosphatasia (HPP). How long did it take for me to say yes? Not very long at all! We in the Genetics and Metabolic community in Manitoba were only too aware of the enormous burden that this serious, debilitating metabolic bone disorder had on infants, children, adolescents and adults and their families in our province and beyond. We were very fortunate to receive incredible support to proceed from all levels of Canadian government as well, in Manitoba, from the Winnipeg Regional Health Authority, University of Manitoba and the Children's Hospital Research Institute of Manitoba and we were off! We developed our clinical research team in Winnipeg and the next few years were very exciting ones as evidence accumulated that ENB-0040 was effective in reversing the devastating effects that this disease had on individuals with HPP and the safety profile of ENB-0040 appeared excellent. Subsequently in December 2011 Alexion Pharmaceuticals Inc., based in Cheshire, Connecticut, acquired the development and commercial rights to ENB-0040 (Asfotase Alfa). Now, 4 years later, Asfotase Alfa was recently approved by Health Canada for the treatment of pediatric-onset HPP; that is for those with onset of the signs and symptoms of HPP less than 18 years of age. It will be marketed under the name StrensiqTM. This is undeniably the most significant milestone to date for the treatment of HPP!

The next step following this approval is for an assessment and recommendation regarding funding of StrensiqTM to the Ministers of Health of our provinces and territories by the Common Drug Review (CDR), a program of CADTH (Canadian Agency for Drugs and Technologies in Health). CADTH is a national organization whose stated mission is to provide "Canada's federal, provincial and territorial health care decision makers with credible, impartial advice and evidence-based information about the effectiveness and cost-effectiveness of drugs and other health technologies" and make recommendations regarding funding. Alexion Pharma Canada, in parallel with the CDR process, is also working towards access to therapy in the future.

We in the medical and scientific communities in Canada are excited about moving to this next phase in the treatment of HPP and your Scientific Advisory Board will work closely with all of you to ensure all patients with HPP who are eligible for treatment will have sustainable access to this drug that has shown incredible promise to date! Please see the attached names, short introductory biographies and photographs of your Scientific Advisory Board. More information will be forthcoming highlighting each of the Board members and the diverse expertise each brings to Soft Bones Canada. Much more information about HPP and the Canadian scene will be forthcoming in upcoming newsletters as well as other events planned by Soft Bones Canada. In the interim please email me at cgreenberg@exchange.hsc.mb with any questions you may have. I will either address them myself or ensure they are directed to the person best able to respond. My warmest personal regards,

Cheryl Rockman-Greenberg MDCM
CHAIR, SCIENTIFIC ADVISORY BOARD

MEET THE SCIENTIFIC ADVISORY BOARD

Cheryl Rockman-Greenberg, MDCM, FRCPC, FCCMG

CHAIR, SCIENTIFIC ADVISORY BOARD

Dr. Rockman-Greenberg is a medical geneticist in the Program of Genetics and Metabolism, Winnipeg Regional Health Authority, and Distinguished Professor, Departments of Pediatrics and Child Health & Biochemistry and Medical Genetics, University of Manitoba. She is a clinician scientist in the Children's Hospital Research Institute of Manitoba and has led the enzyme replacement therapy clinical trials of asfotase alfa for the treatment of Hypophosphatasia in Canada.

Distinguished professor, Depts. Of Pediatrics and Child Health & Biochemistry and Medical Genetics, University of Manitoba and the Children's Hospital Research Institute of Manitoba.

Dr. Philippe Crine

Dr. Crine is a former professor in the Department of Biochemistry at the University of Montreal. Dr. Crine was a founder of Enobia Pharma where he served as Chief Scientific Officer and Vice President Research from 2003 until 2012. He is a co-inventor of Asfotase Alfa and was responsible for its preclinical development. He is now a private consultant providing strategic and technological advice to emerging companies developing therapies for rare bone diseases.

Dr. Leanne Ward, MD, FRCP, FAAP

Dr. Ward is an Associate Professor of Pediatrics (Faculties of Medicine and Surgery at the University of Ottawa), the Director of the Pediatric Bone Health Clinical and Research Programs, and the Principal Investigator of a Canada-wide research program evaluating the effect of glucocorticoids on bone health in children with leukemia, rheumatic conditions and nephrotic syndrome (called the STeroid-associated Osteoporosis in the Pediatric Population (STOPP) research program). She has received a number of awards for her work on the effects of chronic childhood illnesses on the developing skeleton, including the Canadian Child Health Clinician Scientist Career Development Award, the Canadian Institutes for Health Research New Investigator Award, the Canadian Child Health Clinician Scientist Career Enhancement Award and most recently, a University of Ottawa Research Chair Award.

Jessica Hartley, BSc(Hons), MS, CGC

Jessica Hartley has been a genetic counsellor in the Program of Genetics and Metabolism at the Health Sciences Center since 2009. She received a Masters of Genetic Counselling degree from Northwestern University in 2008 and was certified in Genetic Counselling from the American Board of Genetic Counselling in 2009. In her current position, Jessica works in pediatric metabolics and is an integral part of the Manitoba newborn screening program. She has a specialized interest in the psychosocial impact of childhood-onset metabolic genetic conditions and multidisciplinary & interprofessional care models. She is currently a Lecturer in the Department of Biochemistry & Medical Genetics, College of Medicine, Faculty of Health Sciences, University of Manitoba.

Dr. Frank Rauch

Dr. Rauch is a pediatrician and a clinician scientist in the Dept of Pediatrics at McGill University. His area of expertise is in metabolic bone diseases including Osteogenesis Imperfecta. He works at the Shriners Hospital for Children in Montreal.

Amy Yakimoski, RN,BN

Amy has worked as the clinical trials coordinator for Asfotase Alfa at the Winnipeg site in the Children's Hospital Research Institute of Manitoba since 2010 and has been instrumental in meticulously meeting the needs of all the HPP patients who have participated in clinical trials.

Dr. Sandra Sirrs, FRCPC

Dr. Sirrs is an adult endocrinologist and is the Medical Director of the Adult Metabolic Disorders Clinic in the Dept of Internal Medicine, University of British Columbia. She specializes in inborn errors of metabolism and, as a clinical trialist, she has been instrumental in the development of clinical practice guidelines for enzyme replacement therapy for a variety of lysosomal storage disorders including Fabry disease.

Mark DeBrincat

Mark DeBrincat is a Physiotherapist who specializes in Orthopaedics and Sports Medicine. He lives out his passion for his work and has a thriving private practise in Winnipeg, Manitoba. For 28 years, his unique methods of Orthopaedic manual therapy, Myofacial techniques, Exercise regimes and Acupuncture expertise have been recognized and well sought after. He has had extensive experience and success in treating patients with HPP and is both pleased and honoured to sit on the Scientific Board.

Dr. Marc D. McKee

Dr. McKee is a professor at McGill University in Montreal in the Faculties of Dentistry and Medicine. He received his B.Sc. and Ph.D. degrees from McGill University in cell biology, and after a postdoctoral fellowship in the Department of Orthopedic Surgery at Harvard, he held an academic appointment at University of Montreal, after which he moved to McGill University in 1998. Dr. McKee has contributed greatly to our understanding of the causes of dentin and pulp pathology in the teeth of HPP patients.

Dr. Bob Schroth, DMD, PhD

Dr. Schroth is a dentist and a research scientist at the University of Manitoba. He actively researches the relationship between oral health and vitamin D status especially in First Nations children. He has been an active member of the clinical trials team at the University of Manitoba of Asfotase Alfa for the treatment of HPP.

MOVING FORWARD — OUR GOALS FOR 2015-2016

- Develop a patient network connection.
- Develop a health care provider and services referral base.
- Print and distribute uniquely Canadian Soft Bones Canada brochures and publications for patients, caregivers and medical professionals.
- Host educational Webinars.
- Host a national patient meeting.
- Print and distribute newsletters seasonally.
- Continue to assist patients and caregivers in finding quality medical care.
- Continue to provide on-going patient and caregiver support.

SBC's goal for our newsletter, is to provide useful and interesting content relating to issues and topics about HPP in Canada and around the world. Discussion of medical and health topics should never be construed as medical advice. You are advised to contact a health care professional in your area. SBC does not guarantee that the information in the newsletter is complete, or without error, nor do we endorse any particular therapy. We will endeavour to provide a cross-section of information so that you can have an informed dialogue with your health care team, as there are a variety of HPP symptoms and severity differs from patient to patient.

Soft Bones Canada

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