

In This Issue:

- President's Address
- A Note of Thanks
- Submission Request
- Introductions
- Rare Disease Day 2019
- Hoodies For Sale
- Congratulations Dr. Rockman-Greenberg
- Meetings / Conferences
- Board of Directors Meeting
- Scientific Advisory Board—Updates



President's Address

- Linda Toews

Happy New Year from Soft Bones Canada.

This last year has seen some big changes to our organization. Debbie Taillefer stepped down as president and I stepped in to fill very large shoes. We wish Debbie the best as she starts a new phase in her life.



With the change in leadership, we have also added 2 new directors to our Board of Directors. Jennifer Paulson and Eric Neufeld have joined our team. We look forward to their hard work and support for Soft Bones Canada. We have also revamped our

website and will continue to update it so that our HPP families can have access to new and changes in the HPP world.

This fall we were able to get together as a board in Calgary, Alberta. This meeting was a planning session for the next year. Our main focus in 2020 will be creating a dental brochure for dentists and HPP patients/families. This has been a important project for us and one we have been working on for some time. Our SAB has been working with us to create this brochure and we hope to start printing it this spring.

May health and happiness follow you and yours throughout the year to come.



A Note of Thanks

Soft Bones Canada has truly been blessed to have Debbie Taillefer's time, experience and support for many years. Debbie was the founder of the first HPP website in Canada and we thank her for taking on the monumental task of beginning Soft Bones Canada in 2013.



For so many in our HPP community, Debbie has been their lifeline to information and support. Debbie is the first person that I connected with when I was diagnosed. She gave me comfort and help at a time when I needed to connect and educate myself. She was Soft Bones Canada before it began. Debbie encouraged me to become a member of SBC and through her leadership, I believe that I have experienced a great deal of personal and professional growth.

As Debbie moves forward on her own path, I want to thank her both personally and professionally for helping start this organization that has helped so many.

Soft Bones Canada was founded with the intention to support patients and families who are living with HPP and we will do our best to continue the important work that Debbie began here in Canada.

- Jennifer Boin

SUBMISSIONS NEEDED —

Please read!!



Patented
Medicine Prices
Review Board

The Patented Medicine Prices Review Board (PMPRB) released its new draft Guidelines this month and launched a 60-day consultation period with stakeholders and interested members of the public. This consultation follows Health Canada's recent amendments to the Patented Medicines Regulations, which strengthen and modernize Canada's pricing framework for patented drugs so that the PMPRB can better protect Canadian consumers from excessive prices in an era marked by increasingly high cost drugs.

The Guidelines explain the steps taken by staff at the PMPRB in assessing whether a patented drug appears to be priced excessively in Canada. It also explains what information patent-holding pharmaceutical companies must provide to the PMPRB to enable it to make that assessment.

The deadline for providing written submissions to PMPRB.Consultations.CEPMB@pmprb-cepmb.gc.ca is

January 20, 2020.

The PMPRB consultation plan also provides for multiple feedback opportunities and formats.

<http://www.canada.ca/content/dam/pmprb-cepmb/videos/consultations/draft-guidelines/draft-guidelines-en.mp4>

Welcome New Directors!

Eric Neufeld and Jennifer Paulson have volunteered to join SBC's Board of Directors. They come to us with many skills, abilities, and each have a strong connection to the struggles that HPP patients and families live with. We are so excited to have them with us.

Introductions

Eric Neufeld

As we pause for a moment to reflect on the year that has been, I would like to thank all those who have given of their time and effort in continuing the work of Soft Bones Canada. Much has been done to advance the care and community of those living with HPP, and for the Board of Directors it is our desire to do even greater things in 2020.

In my first year as a Director for Soft Bones Canada, I have been privileged to serve with other colleagues who share the same passion about HPP awareness. I have also seen the doors open for others to become involved in this great work. As we look to increase our ability to assist those living with Hypophosphatasia, we also recognize that more help will be needed in this endeavor. I'd like to invite you to join us in this great cause by getting involved in your local communities to raise awareness of HPP.

If there is anything I can do for you, or for more information on what you can do to help, please don't hesitate to contact me.

May you have a Happy and Joyful New Year!

Eric Neufeld

Jennifer Paulson

Jennifer Paulson also joined the Board as one of the new directors of SBC this year. Jennifer has been involved with the SBC community in different ways for several years. She is a parent and caregiver of two young girls with hypophosphatasia living in Alberta. Her older daughter participated in the asfotase alfa trials and receives Strensiq. She started up the Alberta Connect SBC group last year and is excited to be working with the Board.



Rare Disease Awareness— February 28, 2019



As part of her work as a high school teacher, Jennifer Paulson organized a Rare Disease session for the school's Wellness Day Symposium on February 28, 2019, which was also Rare Disease Day. They were very lucky to have two young HPP patients as guest speakers and leaders of the session. Simone (11) and Willem (16) did a fabulous job telling their stories and leading sharing circles.

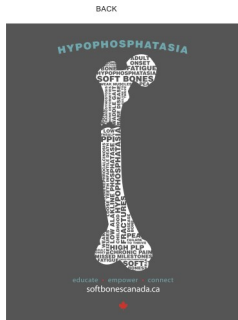
As always, if you have ideas, or would like get involved, please get in touch with us at contactus@softbonescanada.ca

SBC Hoodies

FOR SALE

\$55.00

(taxes and shipping included)



Colour = charcoal (as above)

All adult and youth sizes available.

Order by February 1st, 2020

Visit our website after Jan. 10th at
softbonescanada.ca,

or call

1-204-202-3211

Dr. Cheryl Rockman-Greenberg Inducted into The Order of Canada

Congratulations to Dr. Rockman-Greenberg who will be invested into the Order of Canada. This honour recognizes those Canadians who make extraordinary contributions to the nation.

Dr. Rockman-Greenberg is a well-known and much loved doctor, scientist, caregiver and friend to the HPP community in Canada and the world. She is also the chair of our Scientific Advisory Board. We are thrilled that she is being recognized for her on-going commitment to medical research and patient care for rare disease.

Congratulations





Jennifer Boin with Dr. A. Khan in London, ON

Medical Meetings—London, ON April, 2019

15th Annual Canadian Endocrine Update (CEU) Scientific Meeting—April 26th

National and international faculty presented advances in endocrinology and metabolism. Specifically, the meeting focused on the diagnosis and management of lipid and calcium disorders, diabetes, genetic aspects of endocrine diseases, osteoporosis and hypogonadism.

Aliya Khan, Co-chaired the meeting and presented information about hypophosphatasia during the rare bone symposium which took place on the 26th.

8th Annual Canadian Endocrine Review Course (CERC) April 27th

The focus of this event was for practicing physicians from all fields to prepare trainees for the board exams in Endocrinology and Metabolism or Internal Medicine.

Many thanks to Lisa McGuffin who joined Jennifer Boin to attend this conference. SBC had a table set up with other health organizations and companies.

This was a great opportunity to make connections that we hope will pay dividends as we move forward.

New Dental Brochures for 2020

We look forward to the creation and distribution of an HPP dental brochure in the new year.

Thanks to Dr. Bob Schroth and his colleagues for their work in creating a Canadian publication that focuses on HPP as a dental disease.

The hope is that patients will be able to bring this brochure with them to appointments to assist the dental professionals in understanding HPP and the unique challenges that this disease can present in terms of dental deformity, decay and loss of teeth.

As you may be aware, dental professionals are often the first to notice signs and symptoms of HPP in children's teeth. Naturally, we want to provide as much information as we can to patients, caregivers and medical professionals.



Metabolic Family Workshop

London, ON. May 3, 2019



Dr. Chitra Prasad and
Fernando Boin

Many thanks to Fernando Boin for volunteering to set up and work our display table at this bi-annual workshop put on by the London Health Sciences Centre.

This wonderful event was created to help families make connections, interact with speakers, visit displays and learn ways to care for themselves and their families.

If you know of similar events in your area, please let us know. We'd be happy to spread the news.

RE(Act) Congress 2019/Rare Disease International/CORD Conference May 8-11, 2019

- Linda Toews

This past spring I was able to attend the above conference in Toronto. This was a 5 day conference with the first 2 days with the Re(Act) Congress meeting, followed by the joint hosts of Rare Disease International (RDI) and Canadian Organization of Rare Disorders (CORD). This was an International meeting of scientific leaders, experts and researchers in the rare disease community. We were fortunate to receive a travel grant from CORD in order to cut down on our expenses.

The focus of the first 2 days was with RE(Act) Congress bringing together scientific experts, including researchers and other leaders to present cutting edge information and the exchange of ideas. Some of the sessions covered ideas like Gene Editing and Therapy, Multi-national clinical trials/registries, Indigenous population, and many more.



Canadian Organization
for Rare Disorders

Each one of these sessions included 4-5 speakers, as well as Q&A at the end to exchange ideas and discuss presentations. Though heavy on scientific material, the common thread through the 2 days was the need to connect with patients and families of rare diseases on a local and international level. Many times it was mentioned that the patient/families were needed to make the whole process work and be effective. The trend is to incorporate real world evidence into all the research programs. It was exciting that patients were recognized and needed to be part of the solution. Another theme was to continue to raise the flag on rare diseases. I was able to connect and meet with Canadian, US and European scientists, government officials and patient organizations and discuss how science is changing and impacting rare diseases.

The second half of the conference was hosted by RID(Rare Diseases International) and CORD(Canadian Organization of Rare Disorders) I was able to attend workshops on Voice of Patients, Empowering the Patient and Patient Community. I was able to meet and talk with other patient groups and share the challenges and struggles patient organizations face in pharma, government and medical industries. Each session helped teach what we can do to become one voice for rare diseases. I was able to get tips and ideas of how to have effective dialogues with industry and medical communities.

My takeaway from this conference was new contacts across Canada and tools to move Soft Bones forward in the changing landscape of scientific research. I realized that all countries in the world are trying to get recognition, funding and research on the 7000+ rare diseases identified to date. It will be exciting to see what scientific and technological changes will evolve in the future to cure and treat rare disease.

- Linda Toews

Board of Directors— Calgary

The Board of Directors met in Calgary in late September for a planning meeting.

It was the first time that the new board had met each other in person. The members did some team building at an escape room in Calgary (and successfully escaped) and continued the weekend with a planning meeting. Some of the key issues for 2020 are:

- 1) Fundraising—without an infusion of funds, the organization is in jeopardy
- 2) Education—creation and distribution of a dental brochure
- 3) Patient Engagement— the need to find new ways to get our patients involved in our organization



In November, Jennifer Paulson was invited to the University of Calgary to give a talk to medical students about living with rare disease and the associated challenges and opportunities rare disease can pose for patients.

She spoke to them about her family's journey to reaching an HPP diagnosis, their experience with specialist appointments and assessments, their involvement with the drug trial, and their ongoing monitoring and treatment of symptoms. She was invited as a caregiver and was also able to speak briefly about patient support from the perspective of SBC.



**UNIVERSITY OF
CALGARY**

The feedback was excellent and med students expressed appreciation for learning more about the patient perspective.

If you would like to contribute financially to Soft Bones Canada, please visit our website at:

<http://softbonescanada.ca/donate-now/>

DONATE NOW

Scientific Advisory Board

2019 Report to Soft Bones Canada from the HPP Scientific Advisory Board (SAB)- Dec 14 2019.

The SBC SAB did not officially meet in 2019 but there is lots of important information related to HPP involving Board members and many others working in the HPP field. Areas to report on include:

1) 2019 Manuscripts Published about HPP

A manuscript reporting the 5 year outcome of infants and young children with HPP who participated in the open label trial of asfotase alfa (including children who were enrolled at the Winnipeg site) was published in the Journal of Clinical Endocrinology and Metabolism in 2019. The manuscript is titled **Efficacy and Safety of Asfotase Alfa in Infants and Children Aged ≤5 Years With Hypophosphatasia: a Phase 2 Open-Label Study** by Christine E. Hofmann, MD; Paul Harmatz, MD; Jerry Vockley, MD, PhD; Wolfgang Högl, MD; Hideki Nakayama, MD; Nick Bishop, MD, Gabriel Á. Martos-Moreno, MD, PhD; Scott Moseley, PhD; Kenji P. Fujita MD; Johannes Liese, MD, MSc, Cheryl Rockman-Greenberg, MD, CM, and the ENB-010-10 Study Group. Evidence was presented that, after 5 years of treatment with asfotase alfa, survival, respiratory status and xray findings were significantly improved in most infants and children with hypophosphatasia and that the drug is well- tolerated.

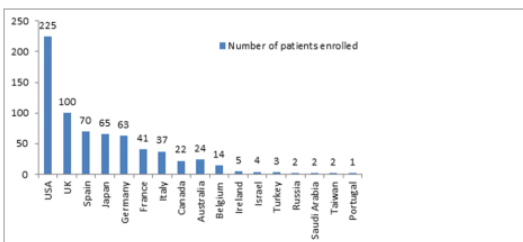
Several other important manuscripts were published by Canadian HPP experts. These include a manuscript by Dr. Aliya Khan (McMaster University, Hamilton) et al entitled **Hypophosphatasia: Canadian Update on Diagnosis and Management in Osteoporosis International 2019** and a manuscript by Dr. Frank Rauch (Shriners Hospital, Montreal) et al entitled **ALPL mutations in adults with rheumatologic disorders and low serum alkaline phosphatase activity in J Bone and Mineral Metabolism 2019**.

These latter 2 manuscripts reflect the growing interest and need for more information about what is the significance, if any, of finding a low alkaline phosphatase level in an adult with and without bone and joint problems. New knowledge is emerging all the time on this topic and in Canada. Very recently a group of medical experts in HPP met in Winnipeg with Alexion Canada to discuss this very point and there are plans moving forward to collect more evidence on the burden of disease in adults with HPP, be they pediatric –onset or adult –onset, the impact of treatment with asfotase alfa, and develop evidence- based guidelines for the treatment of adults.

The guidelines for treatment and monitoring of infants and children with HPP in Canada have recently been posted (Under the guideline tab) on Canada's Garrod Association website: www.garrod.ca. Special guidelines have also been developed for the multidisciplinary management of newborns identified with HPP.

2) Report from the HPP Global Patient Registry

This Registry is an international registry that enrolls consenting patients with HPP. To date there are 680 patients from 17 countries enrolled (see attached graph) including Canada. Given the rarity of HPP and the marked variability from individual to individual in how HPP manifests, there is a need to better understand the natural history of HPP and the full impact of this disorder of the individual and his or her family. This Registry sponsored by Alexion is a tool designed to help us answer these questions. The Registry collects data longitudinally on burden of illness, functional status and disability, and Quality of Life. From the data collected to date we know that diagnostic delay is common in HPP and the burden of illness is high in children and adults.



HPP Global Patient Registry n=680

3) Reimbursement issues

There has been a lot of press lately about new legislation announced by Canada's Patented Medicine Pricing Review Board (PMPRB/CEPMB) of Canada (pmprb-cepmb.gc.ca). The new Federal regulations are to go into effect in 2020 and will impact how new patented drugs, especially as related to drugs for rare diseases, will be assessed and priced in Canada. Some patients and advocacy groups are concerned that these changes will greatly disadvantage access of Canadian children and adults with rare and ultra-rare genetic disorders to new effective treatments. They are concerned that pharmaceutical companies may be less interested both in conducting clinical trials as well as commercializing new rare disease treatments in Canada in the face of these new regulations. Although the HPP SAB has not specifically discussed the impact of this legislation on HPP, we hope that all patients, advocacy groups and physicians continue to collaborate with manufacturers and regulatory agencies on any strategies to improve access to DRD, including new drug pricing models.

- Dr. Cheryl Rockman-Greenberg