JANUARY 2023

SBC NEWSLETTER

Message from the President

Soft Bones Canada would like to extend our warmest holiday wishes to all members of the hypophosphatasia community in Canada and their families!

It has been a quiet year for SBC, though we are delighted to have invited several new patients into the SBC family and strengthened our relationships with other rare disease patient groups in Canada and the US. The end of 2022 saw some major changes to the Board, with Linda Toews offering her resignation as President. As we enter 2023, Linda will remain as a Director, along with myself, Eric Neufeld, and Brent Coakwell, who joined our Board in June of 2021. We are very fortunate to have Brent join us, he offers a wealth of expertise to the team and is a wonderful support to patients seeking to understand the adult perspective of HPP in Canada. We are also very pleased to welcome back our Office Manager, Jessi Neufeld. Jessi is the friendly first point of contact for most patients, and her work for SBC is critical in keeping us organized and at our best!

One of our main focuses in 2022 was completing the HPP dental brochure for patients and practitioners. We are delighted to announce that the brochure is almost complete! Thank you to our Scientific Advisory Board and the rest of the team who contributed with love and attention to this publication. We are in the final stages of editing and we are planning to begin printing the brochures in the coming weeks.

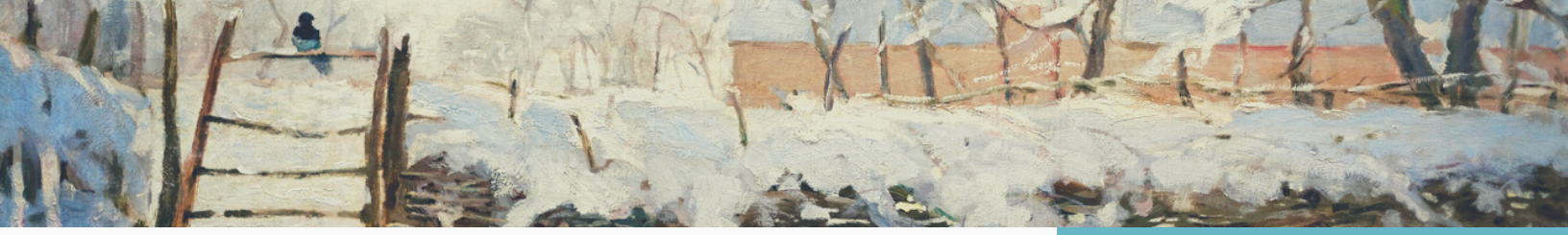
SBC continues to welcome new volunteers to assist with our work to educate, empower, and connect members of the HPP community in Canada. Please see below for more information.

Warmest regards,

Jennifer Paulson
President, Soft Bones Canada

In this issue:

Soft Bones® Canada



Patient Story: Simone

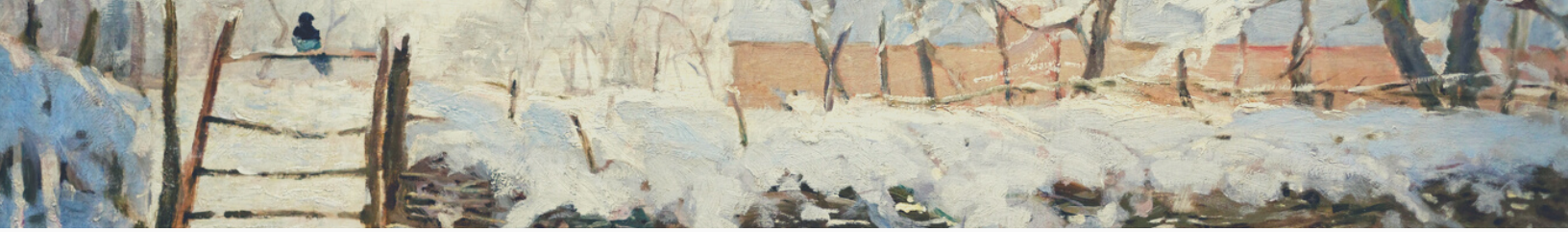
My name is Simone. I have always been a hypophosphatasia patient. I was diagnosed with infantile HPP when I was one year old, and this was mostly because of dental problems. I was lucky that my ALPL mutations were not the serious ones that caused life-threatening complications. But I did have bone deformities, low energy levels, trouble walking and climbing stairs, and I also remember feeling pain in my legs.

I remember going to Winnipeg and having Dr. Greenberg help me and give me the "mosquito medicine" that would help me walk and not have the pain in my legs. This started when I was three years old. I was part of a trial that tested the enzyme replacement therapy that is now called Strensiq. I had lots of fun doing the physio tests and being allowed to run down the hallways of the hospitals. Everyone was so nice. I also got to do lots of other neat things like stay at Ronald McDonald House and be in their fashion show. When Soft Bones Canada started in 2013, I really enjoyed being included in their events and getting to know so many other people who know about HPP. Once I gave a talk at a high school about my experience living with a rare disease, and I shared about how lonely it sometimes is, having a disease that is almost invisible (except for my teeth) and that almost nobody understands.

I am really lucky that I have had such good help from doctors and nurses, and that I was able to get the enzyme replacement medicine that helped heal my bones. I have also had very bad dental problems. I have four dentists and I think I miss school at least twice a month in order to go to see one of them. Last summer I saw a prosthodontist who really made my teeth look awesome, and I was so happy to have him do that. I feel very lucky that the main inconvenience that I have had in my life is missing lots of school and having so much time with doctors. I became really afraid of needles, especially for all the blood tests that I had to have. I went to support programs for needle phobia and also had appointments with a psychologist to help because I was so afraid. I am still quite afraid of the blood tests but I have learned to give my own subcutaneous injections for the Strensiq. For some reason, the injections from the medicine have never frightened me as much. Maybe because I knew they were really helping me live a normal life.

"I FEEL VERY LUCKY THAT THE MAIN INCONVENIENCE THAT I HAVE HAD IN MY LIFE IS MISSING LOTS OF SCHOOL AND HAVING SO MUCH TIME WITH DOCTORS."

- SIMONE



Celebrations from 2022

In addition to joining online celebrations of Rare Disease Day on February 28 and World Hypophosphatasia Day on October 30, SBC participated in a number of other events and meetings in 2022. In June, SBC Directors met with Soft Bones President and Founder Deborah Fowler and Executive Director Denise Goodbar. They shared many exciting ideas and events with us about how Soft Bones in the USA is supporting their HPP community. They invited Canadian patients to explore their website and register as free members of the online community. Patients can join their mailing list to receive their quarterly newsletters, as well as have access to their information and services such as their excellent webinars for HPP patients. Their website includes a busy moderated chat room for patients and caregivers to securely share about their experiences with other members of the HPP community.
Be sure to visit https://softbones.org/ for more information.

SBC was featured in a Health Insight article in September 2022, focusing on bones, muscles, and joints. Author Tania Amardeil interviewed Dr. Leanne Ward and former SBC Director Jennifer Boin to discuss the importance of raising awareness about HPP and rare diseases, the challenges of diagnosis and key aspects of managing the disease.

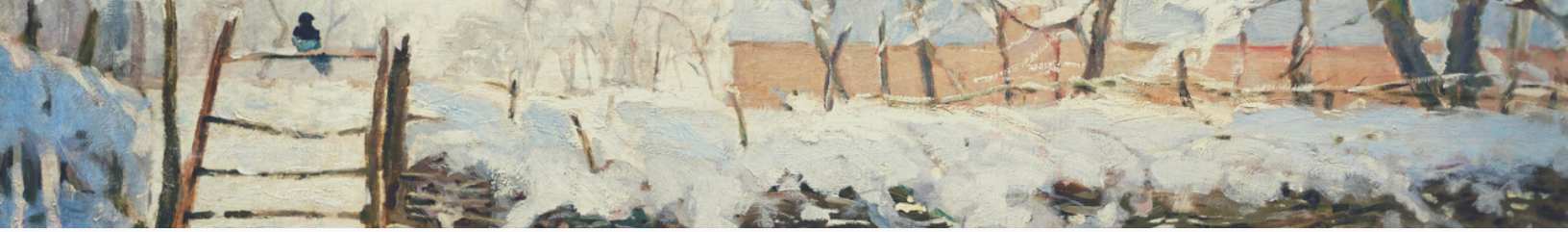
https://www.healthinsight.ca/wellness/bones-muscles-joints/boosting-awareness-of-this-rare-genetic-condition-can-save-live/

Upcoming Events in 2023

Dr. Leanne Ward with CARDS (Canadian Alliance for Rare Disorders of the Skeleton) will be hosting a free webinar on Tuesday, January 31, 2023 at 7pm EDT. All are welcome to attend. This webinar will feature a keynote address by Durhane Wong-Reiger, President and CEO of CORD (Canadian Organization for Rare Disorders) and will focus on the adult perspective on living with rare bone disease. SBC and other bone disease advocacy groups will have representatives sharing their insights and concerns. The webinar is designed to highlight patient perspective and to strengthen the collective voice of patient, caregivers, and patient support groups. Discussion will focus on the unmet needs of adults living with rare bone disorders across Canada. Registration information will be posted in early January on bonescanada.org (click on Past & Upcoming Events → For Patients, Friends, & Families).

Rare Disease Day: February 28, 2023

World HPP Day: October 30, 2023



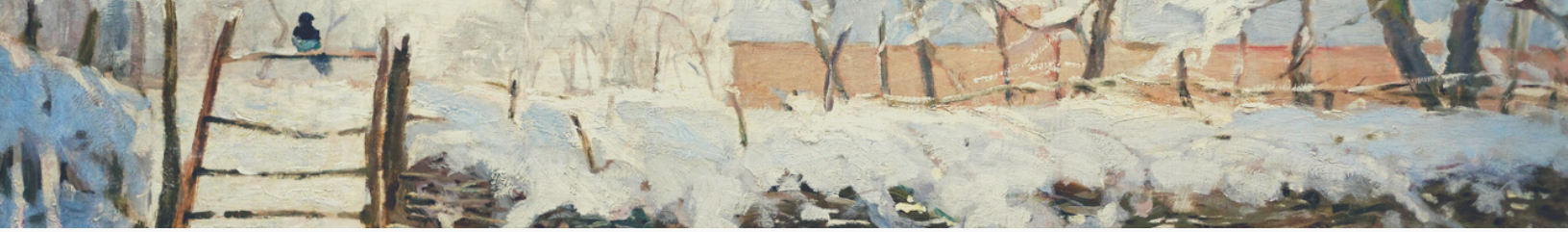
Scientific Advisory Board Report: 2022

Oct 24 2022

Report of the Scientific Advisory Committee to the 2022 Annual Meeting of Soft Bones Canada (SBC)

The Scientific Advisory Committee of Soft Bones Canada (SBC) did not meet during the past year. However, there were many informal discussions between various Board members. The HPP SAC consists of Dr. Cheryl Rock-man-Greenberg (Chair), Dr. Philippe Crine, Dr. Sandra Sirrs, Dr. Marc McKee, Dr. Bob Schroth, Dr. Leanne Ward, Mark Debrincat, Jessica Hartley, Amy Yakimoski and Alie Johnston (Dr. Rockman-Greenberg's research coordinators) and Linda Toews. This year's report is very similar to last year's report.

Dr. Rockman-Greenberg is a member of the Medical Advisory Board of the Alexion- sponsored Global Patient HPP Registry. Amy Yakimoski is the Registry coordinator for the Canada site. As of June 2022, the Registry has 1042 patients enrolled from 71 patient sites. The Registry enrolls HPP individuals who are on enzyme replacement therapy (ERT) or are ERT naïve. The goal of the Registry is to provide Real World Data on HPP. With respect to Canadian stats, 24 patients (adults and children, both on and not on ERT) have signed consents and are actively enrolled in the Registry.No new patients have been enrolled in the past year. HPP Registry Medical Advisory Board met virtually has an upcoming virtual meeting in Dec 2022. For further information on the Registry, please contact Amy Yakimoski at ayakimoski@chrim.ca.



Scientific Advisory Board Report (continued)

RESEARCH UPDATE

• Discussions continue how to expand reimbursement strategies to include Canadian adults with HPP who may meet eligibility criteria. Dr. Rockman- Greenberg has approached CADTH regarding reviewing reimbursement criteria. She has learned that new data must first be formally presented to CADTH which then must undergo appropriate and rigorous review by CADTH. As such she is in discussion with Alexion Canada regarding next steps regarding Alexion filing for a formal reassessment to expand the current reimbursement criteria.

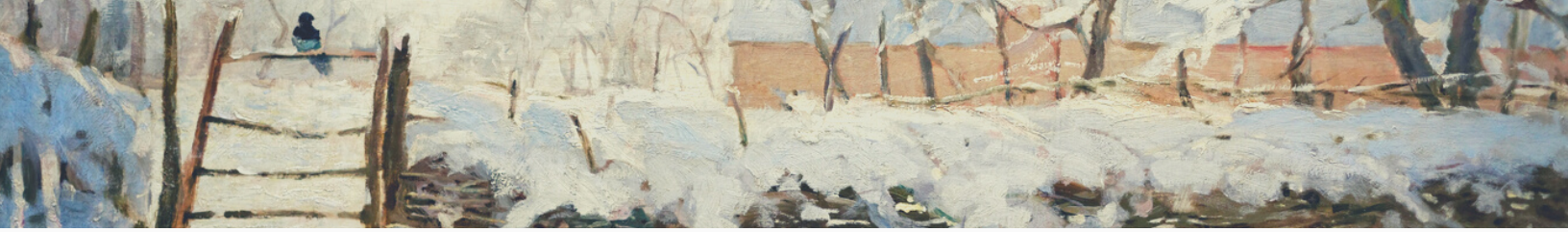
• Dr. Wolfgang Hogler, from Johannes Kepler University in Linz, Austria is the lead principal investigator of a 3 year global research effort to study genetic variants found in the ALPL gene. This study is ongoing. The importance of DNA testing to help establish the diagnosis of HPP is unequivocal. However, often a DNA variant (a variant is a change in the DNA lettering) is found but the significance of the DNA variant is unclear meaning it is uncertain if the variant is actually the cause of the signs and symptoms of the patients (known as a pathogenic variant) or is just a misprint of no significance (known as a benign variant). Classifying an ALPL gene variant as pathogenic or benign is very critical and although there are well established guidelines how to classify variants, there remain many variants whose significance is uncertain. Dr. Hogler's study is called "Functional Testing of Variants of Unclear Significance in the ALPL gene as a cause of Hypophosphatasia" and Dr. Cheryl Rockman-Greenberg is a member of Dr. Hogler's global research consortium. She has submitted data on "variants of uncertain significance" from 2 Canadian patients to this research effort for further testing; 2 additional are pending. The information from this study will be entered into a continuously expanding, publicly available ALPL gene variant database and will greatly enhance our ability to understand which and how gene variants actually cause disease. Please note if a patient with a DNA variant of "unknown significance" consents and wishes to enroll in this study, no DNA sample is sent (such as a cheek swab or blood sample)—only the specific DNA lettering of the ALPL variant found on the initial DNA testing by the Canadian physician involved will be sent to the Consortium molecular laboratory in Linz, Austria for its specialized molecular testing to allow for precise classification of a variant into pathogenic or benign. As DNA testing is critical to the diagnosis of HPP and ultimately to patient care this study will be of great importance. For further information contact Dr. Cheryl Rockman-Greenberg.

• Dr. Aliya Khan from McMaster University and colleagues Dr. Eric Rush (Kansas City) and Dr. Maria Luisa Brandi (Florence, Italy) are leading an International Working Group that has done a systematic review of the literature and is developing diagnostic criteria for pediatric and adult HPP based on scientific evidence. Several publications regarding diagnostic criteria have now been submitted for consideration re publication and are under review.

• BEYOND ERT: There are new research initiatives being led by 2 pharmaceutical companies exploring gene therapy in HPP. These are very exciting times as results of gene therapy in animal models have been very promising and applications are being made to the FDA to explore Phase 1 and 2 trials in humans! Several members of SBC SAB are involved in these ventures.

• RESEARCH PUBLICATIONS: There have been several publications on HPP from the HPP Global Registry on clinical profiles and burden of illness of patients as well as a recent paper by Dr Bob Schroth on Dental Outcomes for Children receiving asfotase alfa for HPP in BONE 152 (2021) 116089.

• The Canadian HPP Clinical Expert Committee is active and receives requests from the provinces to consideration applications for beginning ERT and renewals. Its terms of reference, eligibility criteria for ERT and details of both the application process and the renewal process can be found on the Garrod website www.garrod.ca. This Canadian HPP Clinical Expert Committee is encouraging physicians to provide complete medical information so that the committee can truly make an informed decision.



Scientific Advisory Board Report (continued)

Dr. Rockman-Greenberg wishes to remind SBC Board that at the Children's Hospital Foundation of Manitoba there is a fund designated specifically for HPP patient care, education, research and related initiatives. Any request or queries regarding this fund can be sent to her.

Lastly, the last almost 3 years have been very challenging for everyone and on behalf of the SAB, I extend our hopes that everyone has felt supported and safe during these very difficult times.
Be safe and be well,

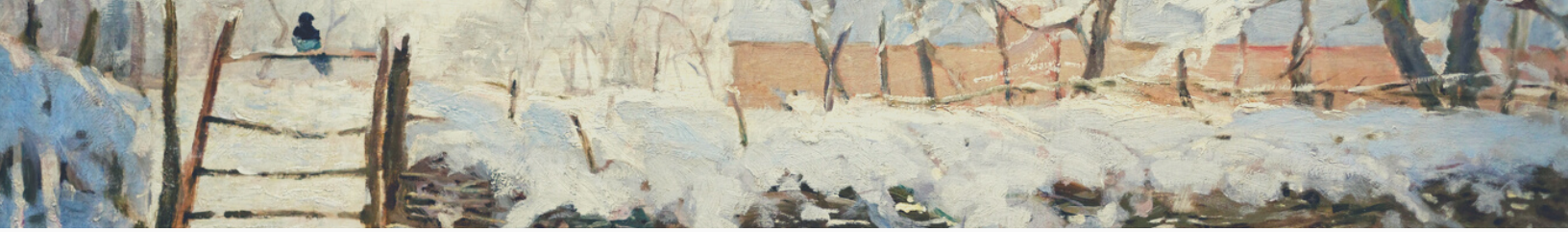
Respectfully submitted

Cheryl Rockman-Greenberg
Chair SBC, SAC

Farewell to Linda Toews as President

Soft Bones Canada has been very well served by our President Linda Toews, who has led our organization with passion and experience for the past three years. In the fall of 2022, Linda announced her resignation from her role as President and she has graciously agreed to continue to volunteer on the Board of Directors. Linda's leadership will be missed and we are grateful to have her continue to serve as a Director. We would like to offer Linda a huge gesture of appreciation on behalf of all HPP families across Canada.

Jennifer Paulson has stepped into the position of President as of October 2022. We wish her the best as she takes on this important role, and works to serve the community with the high standards of integrity and commitment as did our past Presidents, Debbie & Linda.



Call for Volunteers

Soft Bones Canada continues to actively seek volunteers to help us in our work supporting HPP patients in Canada. We are a Canada-wide volunteer-based patient support charity focused on educating, empowering, and connecting HPP patients across the country. We are seeking to expand our volunteer base and accepting expressions of interest at this time.

Opportunities include supporting Provincial Connect groups, fundraising, editorial work, and coordinating community events or HPP-related celebrations. We are also seeking a new member for our Board of Directors. If passionate about the cause, please consider taking on this exciting and important position to support HPP patients in Canada. If interested, please email Jennifer Paulson, President of SBC at jennifer.paulson@softbonescanada.ca

Soft Bones Canada

PO Box 33085 Uplands PO
Lethbridge, AB
T1H 5P2

204 202 3211

WEBSITE DONATE