



JANUARY 2024

# SBC NEWSLETTER

## Message from the President

The SBC Board of Directors would like to extend our warmest holiday wishes to all members of the hypophosphatasia community in Canada!

We have had an exciting year at Soft Bones Canada. First and foremost, we would like to welcome Jeremy Harany to our Board of Directors. Jeremy is a patient living with HPP in Alberta and has been enthusiastically supporting SBC volunteers in all of our endeavours throughout 2023. We are very excited to have him join us and appreciate the skills and experience that he brings to our team. We have included Jeremy's patient story below as a highlight of this New Year's newsletter. Welcome Jeremy!

The Board met in October for the SBC Annual General Meeting. The majority of the excitement was around finalizing our dental brochure (it has now been printed!) as well as new developments regarding access to drug therapies. As announced in our October newsletter, plans are underway for a new enzyme-replacement therapy drug trial from Alexion. We are keenly awaiting more news about location of trial sites and recruitment. There are also new potential developments around expanded reimbursement strategies to provide Canadian adults access to Strensiq. Dr. Rockman-Greenberg, chair of our Scientific Advisory Committee (SAC), has been working with CADTH (Canada's Drug and Health Technology Agency) to have reimbursement criteria reviewed. For more information about this, please see Dr. Rockman-Greenberg's SAC report below. SBC will be writing letters to provincial authorities advocating for criteria that allow adult HPP patients to access drug therapy.

We would like to thank Soft Bones in the United States for continuing to welcome our Canadian patients and share their wealth of resources with our families. We were happy to hear of several Canadian families reaching out to join their Hippo Squad program for children in the HPP community.

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## Message from the President Cont....

We are looking forward to another exciting year in 2024. We hope to be a part of another collaborative webinar next year, and discussions so far have focussed around mental health implications of living with rare disease. We are always taking requests from families, so if there is a topic that you would like to learn more about, please write in and let us know! We hope to continue to expand our social media presence and are looking at some upgrades for our website. Fundraising efforts will also be a priority for us in 2024.

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 Brand  
President, Soft Bones Canada



## Patient Story - Jeremy

Hello, my name is Jeremy. At an early age, I'd lost all my baby teeth which alerted my parents that something was wrong - and (somehow) I was diagnosed with HPP around 1972. I had a specialist until 7 or 8 years of age. I walked with the tell-tale HPP waddle until puberty, but grew to be tall.

As a kid, I was never told to be "extra careful," in fact, quite the opposite - I crashed bicycles, my horse bucked me off more times than I care to admit, and fell out of trees, among other various bumps and bruises. As fate would have it, I had never broken a bone until 6 years ago, and I healed as fast as can be hoped!

In my adult years it has been frustrating finding information about HPP until recently being introduced to Soft Bones Canada. I have been re-diagnosed and finally have proper care. Considering myself lucky, I seem to be limited to dental issues.

I've been amazed and delighted to finally meet other HPP patients and families, and I would love to hear from you!





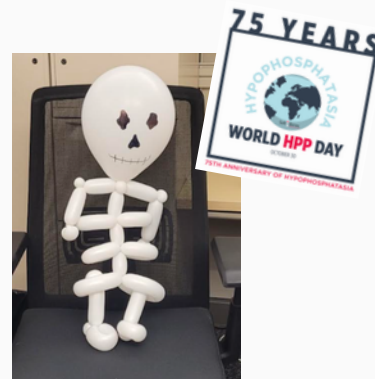


## Highlights from 2023

Our great celebration of 2023 is the official completion of our dental brochure. Thank you to our SAC and everyone who contributed to this publication. We are expecting to print 1,000 copies and distribute to doctors and dentists in early 2024. Keep an eye for digital copies to be uploaded to the website in coming months. We appreciate the generosity of the Deklyn Sawatzky Fund for helping with printing and shipping costs. If you think your local medical or dental office would benefit from some copies of the brochure, please email us to let us know: [contactus@softbonescanada.ca](mailto:contactus@softbonescanada.ca)



World HPP Day was celebrated by the SBC Board in October. Jeremy, Linda, and Jennifer all held celebrations at their workplaces. Jeremy, his colleagues, and their resident balloon artist had some fun connecting with a Halloween theme. The school where Jennifer teaches has a group of students and staff that meet regularly to discuss living with rare diseases and the challenges faced by the families in the RD community. They met for World HPP day as well, and the students campaigned to hold a fundraiser in February to celebrate Rare Disease Day. Plans are underway for that, and it was decided that proceeds will be donated to CORD (Canadian Organization for Rare Disorders).



On the subject of CORD, our Board members were able to attend the in-person CORD conference that was held in Calgary this year. Conversations focussed on the complexities of navigating the “rare disease space” and the challenges of accessing drug therapies for rare disorders. It was wonderful to hear about these new developments in the field and to connect with representatives from other rare disease communities and from industry.

As a final and very exciting highlight from 2023, SBC has also learned of a Grade 7 student named Aaryan who completed a science fair project last year on genetic editing and possibilities for gene therapies in treating hypophosphatasia! He was awarded a bronze medal at the Calgary City Science Fair where he competed against students from all across the city. Congratulations, Aaryan, and thank you for helping raise awareness about HPP!





## CORD Fall Conference

Our Board of Directors attended the CORD (Canadian Organization for Rare Disorders) Fall Conference in Calgary in November 2023. These two days were jam packed with presentations, patient stories, and industry updates about the Rare Disease space. Jeremy and Linda were able to connect with patient group representatives, medical professionals, and industry leaders, and hear where the pathways of treatment and care are headed. Though the focus was rare diseases in general they were able to take away lots of information. Some highlights included:

- Presentation from Health Canada focusing on the new Drug Strategy. The Canadian government has committed to 1.5 billion dollars over the next 3 years. To many, this isn't a cost but rather an investment.
- Importance of patient registries and the impact these can have on supporting new drugs for treatment. The need for a global community committed to have all patients receive the best possible care and newest treatment options. How to provide care for all patients across Canada, and improve diagnosis and medical access to areas outside major cities in Canada: stop the Postal Code Lottery.
- Much focus was on the drug approval and funding pathways leading from initial Phase 1 drug trial to approval for use in patients. Industry and advocacy groups are calling for an accelerated review in Canada that will speed up access to important drug therapies. Jeremy and Linda were able to participate in a group discussion of a rare disease and what a pathway would look like for an unmet need.
- Exciting fronts on cell and gene therapies were presented as well as the new program Canadian Rare Disease Network – “One Child Every Child” to launch in 2024.
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Jeremy and Linda also heard, time and time again, the need for data - more data; lots of data! Yet another key takeaway was the need for attention to mental health and how to better support patients in this facet.

Click on the links below for more information on rare disease and the current landscape:

- Canadian Organizations for Rare Disorders: <https://www.raredisorders.ca/>
- Canada's Drug and Health Technology Agency (CADTH): <https://www.cadth.ca>
- iRARE / Inform Rare Centres: <https://rqmo.org/rare-disease-information-and-resource-centre/>
- INFORM Rare: <https://www.informrare.ca/>
- Care 4 Rare Canada: <https://www.care4rare.ca/>



Jeremy Harany,  
Linda Toews,  
Jennifer Paulson  
Brand at  
CORD in November.

Thank you to CORD for offering SBC a grant that supported our attendance at this conference.



## **Scientific Advisory Committee Report: 2023**

### **Report of the Scientific Advisory Committee to the 2023 Annual Meeting of Soft Bones Canada (SBC)**

The Scientific Advisory Committee (SAC) of Soft Bones Canada (SBC) met one time early in 2023. However, there were many informal discussions between various Board members. The HPP SAC consists of Dr. Cheryl Rockman-Greenberg (Chair), Dr. Sandra Sirrs, Dr. Marc McKee, Dr. Bob Schroth, Dr. Leanne Ward, Dr. Frank Rauch, Mark DeBrincat, Jessica Hartley, Amy Yakimoski and Alie Johnston (Dr. Rockman-Greenberg's research coordinators) and President of SBC. We mourn the loss of our esteemed SAC colleague and friend, Dr. Philippe Crine, who passed away in June 2023.

Dr. Rockman-Greenberg is a member of the Medical Advisory Board of the Alexion-sponsored Global Patient HPP Registry. Amy Yakimoski is the HPP Registry coordinator for the Canada site. The Registry enrolls HPP individuals who are on enzyme replacement therapy (ERT) or who are ERT naïve. The goal of the Registry is to provide Real World Data on HPP. Globally there are 1308 patients enrolled in 12 countries with 68 sites. With respect to Canadian stats, 31 patients (adults and children, both on and not on ERT) have been enrolled and 20 patients are currently active. No new patients have been enrolled in the past year. HPP Registry Scientific Advisory Board met in person in Boston on May 26 2023 and has an upcoming virtual Investigator meeting in Nov 2023. Since 2015, the HPP Registry has published 6 manuscripts- 3 in the past year- and presented 12 abstracts at scientific conferences. The recent publications on HPP from the HPP Global Registry are on clinical profiles of HPP, burden of illness of patients and the spectrum of variants found in the ALPL gene. For further information on the Registry, please see website [www.hppregistry.com](http://www.hppregistry.com) or contact Amy Yakimoski at [ayakimoski@chrom.ca](mailto:ayakimoski@chrom.ca).

Discussions continue how to expand reimbursement strategies to include Canadian adults with HPP who may meet eligibility criteria. Last year Dr. Rockman-Greenberg had approached CADTH regarding reviewing reimbursement criteria. She had learned that new data must first be formally presented to CADTH, which then must undergo appropriate and rigorous review by CADTH but there are other options as well. Alexion Canada has held two meetings of Canadian HPP expert physicians this past year to discuss increasing access to ERT in adults with pediatric-onset HPP. The latest meeting in Oct 2023 discussed the newly approved UK NICE criteria that now reimburses ERT for adults





## Scientific Advisory Committee Report (continued)

With pediatric-onset HPP. The latest meeting in Oct 2023 discussed the newly approved UK NICE criteria that now reimburses ERT for adults with pediatric-onset HPP if 2 of the 3 following criteria are met. The Canadian medical experts at this October 2023 meeting discussed and endorsed the UK NICE criteria with a few modifications and next steps to try to expand the current reimbursement criteria in Canada for adults with pediatric-onset HPP are being discussed, including seeking formal input from SBC and from adult patients with HPP. The UK NICE criteria for the treatment of adults with pediatric-onset HPP include AT LEAST TWO OF THE FOLLOWING:

- Limited mobility assessed by a specialist using the modified Bleck Ambulation Efficiency Score (mobility score) and a Bleck score between 1 and 6 OR
- Current fractures with a history of non-traumatic, recurring or non-or poorly healing fractures OR
- Continuing or recurring musculoskeletal pain that affects daily activities which: affects quality of life and has not gotten better with 2 different types of painkillers that have been recommended by a national pain specialist.

Dr. Wolfgang Hogler, from Johannes Kepler University in Linz, Austria is the lead principal investigator of the global research consortium to study genetic variants found in the ALPL gene. 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. Variants are documented in this continuously expanding, publicly available ALPL gene variant database. The database greatly enhances our ability to understand which and how gene variants actually cause disease. We have recently learned that ongoing funding for this international consortium beyond April 2024, which evaluates the significance of variants identified in ALPL, is in jeopardy. The importance of DNA testing and of this Consortium to help establish the diagnosis of HPP and to inform clinical management is unequivocal. 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.



## **Scientific Advisory Committee Report (continued)**

As DNA testing is critical to the diagnosis of HPP and ultimately to patient care this study will be of great importance. SBC has kindly written a letter to Alexion AstraZeneca Rare Disease strongly advocating for continued funding on this Consortium. This has been greatly appreciated.

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. Two publications regarding diagnostic criteria are now In Press!!

BEYOND ERT: There are new research initiatives being led by two 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. No new recent information is available.

The Canadian HPP Clinical Expert Committee is active and receives requests from the provinces to consider 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](http://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.

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 4 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

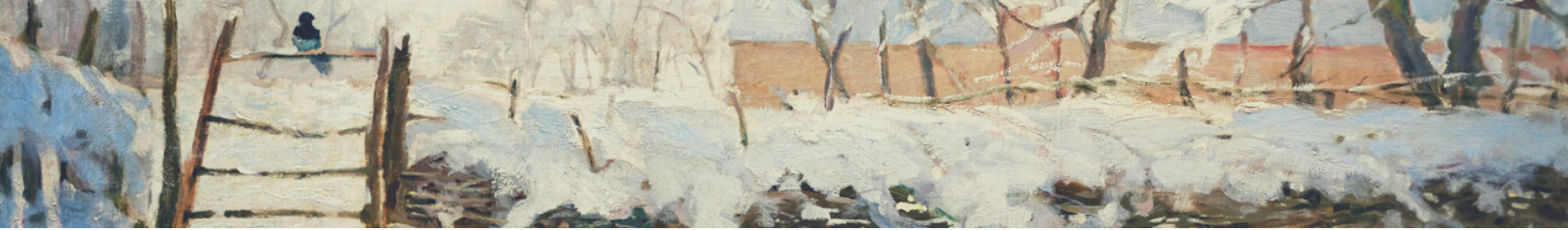


# THANK YOU!

## Thank you to Our Donors

We would like to take this opportunity to thank our generous donors for their financial support this year. SBC depends on contributions from the community in order to continue operating and supporting HPP patients in Canada. Your support is sincerely appreciated!





## 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](mailto:jennifer.paulson@softbonescanada.ca)

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