APRIL 2023

SBC NEWSLETTER

Message from the President

Springtime greetings to all SBC families,

It has been an exciting season at SBC with lots of activity!

I would like to begin by offering our sincere appreciation to Brent Coakwell, who has served as a Director for SBC through a challenging period. We were fortunate to have Brent share his passion and expertise with us over the past two years. In December 2022, Brent resigned from his position as Director and we would like to offer him all best wishes in future ventures. We are happy to share Brent's patient story with you in this issue.

We are also very pleased to announce that we have been joined by a new Board member. Welcome to Jeremy Harany! Jeremy lives in Edmonton, AB, and was diagnosed with hypophosphatasia (HPP) at an early age. He brings insights and experiences that can directly support our adults living with HPP as they manage symptoms and navigate the health-care system. We are glad to have Jeremy working with us to support the HPP community in Canada and his experience is a great complement to the team.

There has been tremendous HPP-related activity lately, a welcome change after the very quiet times of the past couple of years. The Scientific Advisory Board met in February for a lively discussion. Many ideas were shared about the work of SBC including some excellent thoughts around strengthening our patient community, educational webinars, our social media presence, and of course, our beloved dental brochure. See below for information on our upcoming June webinar with CARDS, one exciting event that resulted from that meeting.

SBC Directors have been busy! A planning retreat was held in Calgary back in March where Directors met together in person for the first time in almost four years. Since that meeting, we have been reaching out to our Provincial Connect patients. If you have not heard from one of us already, please expect a call or an email in the next couple of weeks. We like to check in personally with our patients at least once a year, but please remember, you are welcome and encouraged to email us at any time if you have questions or concerns about HPP.

In this issue we will share a Rare Disease Day event that took place in a Calgary school. We are also including information about the Global HPP registry and our upcoming webinar with CARDS. Last but not least, we are very excited to also share information with you about a new HPP drug trial that has a potential research site in Calgary. Researchers for that trial are in the preliminary stages of determining patient interest and availability so please consider filling out their anonymous survey, included below.

Regards,
Jennifer Paulson Brand
President, Soft Bones Canada

In this issue:

Message from the President
PAGE 1
Patient Story: Brent
PAGE 2
Rare Disease Day 2023
PAGE 3
New Drug Trial
PAGE 4
New Federal Funding Announced for Rare Diseases
PAGE 4
"The HPP Registry"
PAGE 5
Upcoming events
PAGE 5
How to Help
PAGE 6

Soft Bones Canada

Patient Story: Brent

My HPP story began as a four-year old child not meeting milestones for physical development. I was having delays and difficulties with walking and gait, and I was experiencing dental abnormalities. I lost teeth and had pain in my legs and wrists, and I spent many years at Sick Kids Hospital in Toronto to support ongoing treatments. Over the course of my life I have had multiple surgeries on all limbs and many, many dental procedures to manage symptoms and deal with complications.

Along with my many challenges, I have also had many successes. I would not be the person I am today without the challenges of HPP. I am blessed to have been married to my wonderful wife Tania for 25 years and have two children: A.J. (22) and Andrew (19). Neither of my children have HPP but both are carriers. I have enjoyed a very fulfilling career in education leadership and also worked as a professional chef.

About five years ago I suffered a bad fall and broke three limbs at the same time. Shortly after that, I was able to access the enzyme replacement therapy Strensiq through my private benefits. Being on this therapy has improved my quality of life by reducing pain levels and increasing mobility significantly. I do find the ongoing approval process to be stressful and challenging as there is always a fear that my insurance coverage will change or that I will no longer qualify. Strensiq for adults is not covered by the provincial drug plan in Ontario.

> “ALONG WITH MY MANY CHALLENGES, I HAVE ALSO HAD MANY SUCCESSES. I WOULD NOT BE THE PERSON I AM TODAY WITHOUT THE CHALLENGES OF HPP.”
>
> - BRENT

Rare Disease Day 2023

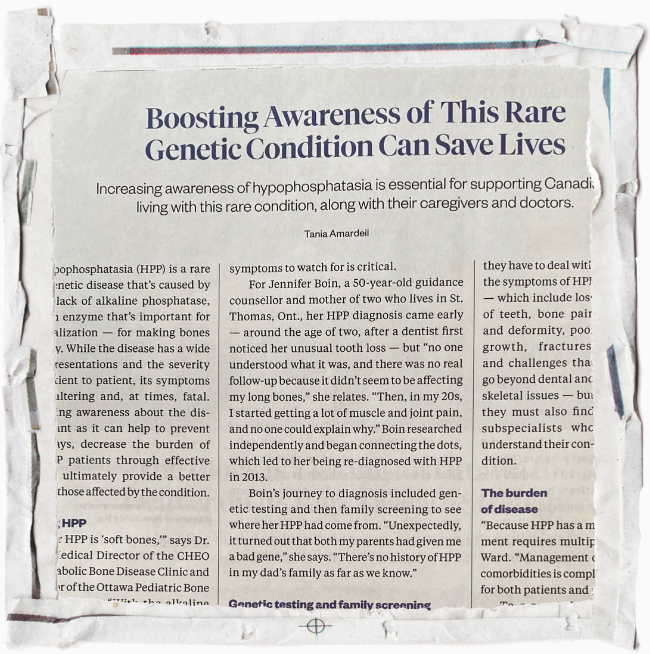
On February 28, students and staff gathered at the large middle school where Jennifer teaches to celebrate the journey of living with rare disease. Teachers who are part of rare disease families led the session where twenty-nine students and five staff members enjoyed a lively lunch meeting together. Everyone introduced themselves and shared about their interest in rare diseases: types of rare disease, prevalence, diagnoses, treatments, psychological aspects, etc. Students were especially keen to hear about careers in rare disease research and treatment. There was so much interest, students requested another opportunity to come together and keep the discussion going. This will be held in May.

If you participated in a Rare Disease Day Celebration, please send us your stories!

HPP Featured in Maclean's Magazine

In our previous newsletter we shared a Health Insight article (September 2022) that featured HPP and included a few words from SBC regulars.

We were delighted to find that this article was reprinted as part of a Rare Disease Special Interest Section in the March 2023 issue of Maclean's magazine.



Boosting Awareness of This Rare Genetic Condition Can Save Lives

Increasing awareness of hypophosphatasia is essential for supporting Canadi
living with this rare condition, along with their caregivers and doctors.

Tania Amardeil

pophosphatasia (HPP) is a rare
netic disease that's caused by
lack of alkaline phosphatase,
enzyme that's important for
alization — for making bones
y. While the disease has a wide
resentations and the severity
ient to patient, its symptoms
altering and, at times, fatal.
ng awareness about the dis-
nt as it can help to prevent
ys, decrease the burden of
P patients through effective
ultimately provide a better
those affected by the condition.

HPP

HPP is 'soft bones,'" says Dr.
edical Director of the CHEO
abolic Bone Disease Clinic and
r of the Ottawa Pediatric Bone

symptoms to watch for is critical.

For Jennifer Boin, a 50-year-old guidance counsellor and mother of two who lives in St. Thomas, Ont., her HPP diagnosis came early — around the age of two, after a dentist first noticed her unusual tooth loss — but "no one understood what it was, and there was no real follow-up because it didn't seem to be affecting my long bones," she relates. "Then, in my 20s, I started getting a lot of muscle and joint pain, and no one could explain why." Boin researched independently and began connecting the dots, which led to her being re-diagnosed with HPP in 2013.

Boin's journey to diagnosis included genetic testing and then family screening to see where her HPP had come from. "Unexpectedly, it turned out that both my parents had given me a bad gene," she says. "There's no history of HPP in my dad's family as far as we know."

Genetic testing and family screening

they have to deal wit
the symptoms of HP
— which include los
of teeth, bone pai
and deformity, poo
growth, fractures
and challenges tha
go beyond dental an
skeletal issues — bu
they must also fin
subspecialists wh
understand their condition.

The burden of disease

"Because HPP has a m
ment requires multip
Ward. "Management
comorbidities is compl
for both patients and

New Drug Trial

The M.A.G.I.C. Clinic in Calgary is being considered as a prospective site for a clinical research trial testing a new treatment for HPP. The trial is anticipated to begin in late 2023 or early 2024. Cohorts of different age groups are being considered, including adults. At this time, the clinic is seeking to determine patient interest to further assess the suitability of the site. Patients or caregivers are invited to complete the following anonymous survey to help the M.A.G.I.C. Clinic gather information about patient interest. More information to be found by following this link to the survey: https://hushforms.com/magiccalgary-6028

For information on the current trials being conducted at the M.A.G.I.C. Clinic please see their website: CLINICAL TRIALS | M.A.G.I.C Clinic (magiccalgary.ca)

SBC will update the community when more information about this drug trial becomes available for public knowledge.

New Federal Funding Announced for Rare Diseases

It is with great pleasure and excitement that we can announce recognition from the federal government how difficult and expensive it can be for patients to access treatment for rare diseases. Our friends at CORD (among others) have been hard at work, and were directly involved in this process. Currently, the federal government is discussing “up to $1.4 billion over three years to provinces and territories through bilateral agreements.”

INVESTMENTS TO SUPPORT ACCESS TO DRUGS FOR RARE DISEASES - CANADA.CA

"The HPP Registry"

SBC would like to share more information about the HPP Natural History Registry, also known as the Global HPP Registry, that is helping to advance understanding of HPP worldwide. Several of our patients have been asking about it, and many of us contribute information to the registry already. As some of you may know, current ongoing research about HPP is limited, and the registry represents an important gathering of information to help researchers better understand many aspects of HPP: the burden of the disease, diagnostic methods, variability in presentation between patients, treatment, and quality of life. The information below about “The HPP Registry” has been provided by SBC’s Scientific Advisory Board.

The Global HPP Registry is an observational, prospective, multinational study initiated in 2014 (NCT02306720; EUPAS13514). The Registry is sponsored by Alexion, AstraZeneca Rare Disease (Boston, MA, USA) and is overseen by an international scientific advisory board of clinical experts, including employees of Alexion.
The HPP Registry team recently met with the HPP investigators and the study teams globally and we learned about:

- ***The current status of the Global HPP Registry**. As of December 12, 2022, the Global HPP Registry contains 1204 patients, representing 12 countries with 1206 patients enrolled including 676 adults. Canada has enrolled 31 patients with HPP.*

- ***Registry Achievements**. Over the past year several manuscripts have been accepted or are in final preparation on the burden of illness in adults with HPP, growth of children with HPP, and the burden of rheumatologic (non-skeletal) manifestations in patients with HPP.*

- ***The ALPL Gene Variant Database and Consortium** is very active under the leadership of Dr. Wolfgang Hogler from Johannes Kepler University in Linz, Austria, and contains information on hundreds of ALPL variants described to date.*

Upcoming Events

Dr. Leanne Ward of CARDS (Canadian Alliance for Rare Disorders of the Skeleton) will be hosting a new educational webinar later this spring. Dr. Ward has invited SBC to co-host this event June 20th, 2023 at 7 EDT. The topic of the webinar will be: "The psychological effects of living with a rare bone disease on patients and families."

All are invited to attend this free session. Registration information coming soon at: bonescanada.org (Past & Upcoming Events → For Patients, Friends and Families).

How to Help!

- Join our community! It's as easy as sharing your experiences with others from the SBC by joining our Facebook page or following us on Instagram.
- Let us know if you'd like to submit a patient story!
- Thank you to our regular donors - your generous contributions make our work possible!
- Seeking new volunteers - you can directly assist us in our work to spread awareness. Please email us at contactus@softbonescanada.ca if you are interested in joining our team.

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