

# THE BARE BONES

Official Newsletter of Soft Bones, Inc., The US Hypophosphatasia Foundation

## Empowering Impact: World HPP Day and Cannonball 5K

This year, World HPP Day coincided with the 75th anniversary of Canadian pediatrician Dr. John C. Rathbun's discovery of hypophosphatasia. This year's theme was "Let's Make Noise about HPP".

Our HPP community created momentum by raising awareness of HPP globally in many ways. Like participating in the Cannonball 5K, sharing stories, and more.

With more than 50 registrants, the HPP community came together to bring global awareness to hypophosphatasia through our 5K. We would like to extend a huge thank you to all of the participants. Special thanks to Alexion AstraZeneca Rare Disease for supporting this event and our HPP community!

Many of our ongoing programs, events, and resources would not be possible without your support. We are grateful for the many patients, families, and caregivers who dedicate their lives and time to building our HPP community and sharing stories and resources. If you haven't already, we invite you to make a meaningful impact by donating to Soft Bones.



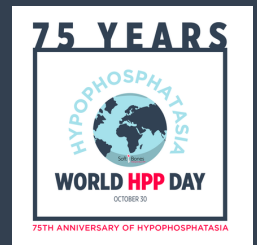
## Sustain Our Mission. Make an End-Of-Year Donation Today!

You Can Make a Difference in the Life of an HPP Patient. Join us in celebrating the stories of strength and resilience in this year's [video](#) as we proudly showcase 12 individuals featured as our Patients of the Month. Together, we can continue building a more supportive and informed HPP community.



### IN THIS ISSUE:

- World HPP Day Celebration
- 2023 Research Grant Winner
- Evin King Spreads Awareness
- Holiday Fundraising Corner



# UPCOMING EVENTS



## 2024 National Patient Meeting July 12-13, 2024

HPP patients and families are invited to the 2024 National Patient Meeting on July 12-13 in Mason, Ohio (near Cincinnati). Join us as we gather to learn more about HPP, share experiences, exchange insights, and foster community. Engage with leading healthcare experts and strengthen your support network. Don't miss this opportunity to meet others living with HPP, their families, and caregivers from all over the country. Check out the [AGENDA](#) for an overview of the planned events and speakers. Registration is now open and travel grants are available. Register [here](#).

## Old School Bagel

COFFEE • BREAKFAST • LUNCH

Old School Bagel - Piedmont  
12000 Northwest Expy Suite A,  
Yukon, OK 73099.

## Central Region Meeting January 13, 2024

Do you live in Oklahoma, Colorado, Arkansas, Kansas, Missouri, North Dakota, South Dakota, Nebraska or Wyoming?

If so, please join Central Region Leads, Nellie Sanders and Casey Anne Johnson for refreshments on Saturday, January 13th, 2024 from 10-12 PM (Central). All Central Region members and families are warmly invited to connect, come together, understand challenges, and learn more about your local HPP support system! Refreshments will be served. Mileage reimbursement is available.

Registration is required and will close on January 5, 2024. Register [here](#).



## South Central Region Meeting March 2, 2024

Do you live in Texas or Louisiana?

If yes, please join South Central Region Leads, Cami Rush and Cassandra Self on Saturday, March 2, 2024 at Texas A&M University. Dr. Dana Gaddy and Dr. Larry Suva will present their research and take us to visit the HPP sheep. Lunch will be served. Registration will open soon.



## HPP AND ME Community Chat January 8, 2024, at 1 PM Eastern

Meet the faces behind the names, talk about HPP or just life in general, and connect with others! Register [here](#)!



HPP AND ME

## HPP AND ME Zoom Tutorial January 8, 2024, at 8 PM Eastern

Learn more about the HPP Community, connect with others, and learn how to navigate HPP AND ME. Register [here](#)!



# HPP Happenings

## 15th Annual Golf Classic

The 15th Annual Soft Bones Golf Classic, held on October 2nd at Somerset Hills Country Club in Bernardsville, NJ, was our largest fundraiser this year. This year's golf classic was another successful event, drawing in 112 golfers who joined us for a beautiful fall day. Sponsors, donors, patients, and participants enjoyed playing on the course while helping raise critical funds for our organization.

A special thank you to our Tournament Sponsor, Atlantic Health System. In addition, a huge thank you to our other sponsors: The Fowler Family Charitable Foundation, Gates & Mary Ellen Hawn, Donnelly Construction, College Mind Counseling - Kristen Tarantola, William F. Jones, DMD, and Ted Walsh - Jupiter Capital Partners LLC. Following the day's triumphs, all participants received takeaway dinners from Perottis in

Bernardsville, NJ. We are grateful for the loyal golfers who turn out year after year to support our mission. This event remains a cornerstone in advancing our mission. We hope to see you at our next golf outing on September 23, 2024, at Somerset Hills Country Club!





## Soft Bones Presents at the OI Foundation Conference

On October 19-20, Deborah Fowler (President of Soft Bones) and Denise Goodbar (Executive Director) traveled to Arlington, VA to participate in a meeting hosted by the OI Foundation titled, "Somewhere to Go for Adults with Childhood-Onset Rare Diseases: A Conversation About How We Can Fill Gaps in Care."

Deb presented in a session titled, "Centers of Excellence (COEs) - Will They Work for Our Community?" She shared the main takeaways from our COE research initiative, which can be found [here](#) (page 9).

During the meeting, attendees worked to address the issues adult patients with rare diseases face as they navigate the healthcare system. Topics covered included examining current rare disease care infrastructure, the role of technology in providing quality care, payment model issues,

patient engagement issues, and current care models such as from the Centers of Excellence and existing consortium and network models. We are grateful to the OI Foundation for including us in this meeting and look forward to continuing this important conversation.



## Evin King Spreads HPP Awareness

In this edition, we are excited to feature Evin King. This past fall, he completed his Senior Capstone project where he presented his research paper titled, *More research needs to be put into rare disease*. To coincide with his research, he also wrote a book geared towards children who have the disease.

Motivated by a lack of relatable content besides scientific journals, he wanted to raise awareness of HPP and share his story. He commented, "This project meant a lot to me. When I was diagnosed as a young child, I did not have any resources that were geared towards my age. I wanted to make a book that would help other children with HPP understand and know my story with HPP".

He believes that creating content and sharing his personal experience will help to support funding for HPP. In presenting his project, Evin successfully brought attention to HPP, which prompted great questions from the judges who had never heard of HPP. We are very proud of Evin for all he has done to amplify the voices of individuals living with HPP.

You can read more about Evin's HPP journey [here](#). Way to go, Evin!

“ I want to educate younger patients with HPP so they do not have to focus on the negatives of the illness but rather illustrate that there is always hope. ”







## Soft Bones Names Claire Stenhouse, Ph.D. as the 2023 Maher Family Annual Hypophosphatasia Research Grant Recipient

Soft Bones Inc. awarded its 2023 research grant to Claire Stenhouse, Ph.D., an assistant professor in the Department of Animal Science, Center for Reproductive Biology and Health, Huck Institutes for the Life Sciences at Pennsylvania State University. The \$25,000 grant will support Dr. Stenhouse's studies to understand the respiratory complications of hypophosphatasia (HPP) in newborns and infants severely affected by HPP.

Soft Bones funds research to understand HPP and the goals to find a cure. The foundation has awarded over \$500,000 in research grants since 2014, supporting HPP investigators through this Maher Family Annual Hypophosphatasia Research Grant. Read the full release [here](#).



## Research Corner

### 2023 Second International Meeting Summary and Videos on Demand for Clinicians and Scientific Researchers

Soft Bones held its Second International Scientific Meeting convening clinicians, physician-scientists, basic science researchers, and industry representatives from around the globe to understand and improve the treatment of hypophosphatasia (HPP). Attendees shared the latest advances and discussed ongoing and future research concerning this rare and complex inborn error of metabolism that manifests the broadest range of severity of all skeletal diseases.

For a summary of the meeting, a link to the published abstracts in the journal JBMR Plus, and more, click [here](#)!



## CoRDS International HPP Contact Registry

The CoRDS International HPP Contact Registry was designed to be an accessible database where every patient with HPP registers to be informed about research opportunities. It allows researchers to alert patients about research opportunities without compromising patient information.

There are many different HPP registries in place and data is "owned" by different stakeholders. For example, Alexion has an HPP registry with data collected by physicians to understand lab readings, side effects, and other ways of tracking patient progress on medication. Other registries exist around mutations and symptoms to help the HPP medical community better understand the connection between genotype and phenotype.

Soft Bones has partnered with the non-profit Coordination of Rare Diseases at Sanford (CoRDS) which allows YOU to be in charge of your data. Your information may be removed at any time. CoRDS is the largest, free international rare disease registry for individuals diagnosed with hypophosphatasia (HPP) and carriers of HPP.

Please consider signing up today to help advance research for hypophosphatasia. The registry is open to anyone diagnosed with HPP, those who are carriers, those who are undiagnosed, or suspect they may have a rare metabolic disease.

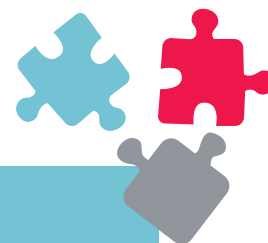
For more information, contact CoRDS at [cords@sanfordhealth.org](mailto:cords@sanfordhealth.org) or (877) 658-9192.

You can also access the [CoRDS Participant Brochure](#), [CoRDS Registration Tip Sheet](#), and [CoRDS Q&A](#). Once ready, you may register using the link below, or call the CoRDS office directly.

Enroll [here!](#)



**You are the missing piece...**



### Invitae Skeletal Dysplasia Genetic Test

Invitae recently launched the Discover Dysplasias Program. It is a no-charge genetic testing for individuals with skeletal dysplasia. Skeletal dysplasia is described as a group of disorders characterized by abnormal bone growth and development. Mutation testing is critical in better understanding the disease and how it impacts families in the future.

#### Do you qualify?

To qualify for this program, patients must be two years or older in the US and Canada with signs or symptoms consistent with a diagnosis of skeletal dysplasia.

Here is the [link](#) to the flyer. You can list eligibility criteria. Remember, the test can only be ordered through a physician. Invitae also offers family variant testing up to 150 days from the report date for any blood relatives (for a positive result).

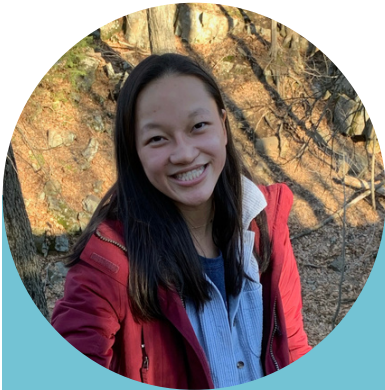




# Employee Spotlight



## Meet Margaret Robb



**COMMUNICATIONS  
MANAGER AND  
PATIENT/CAREGIVER  
NAVIGATOR**

Hello! My name is Margaret Robb. I am the Communications Manager and Patient/Caregiver Navigator. What drew me to Soft Bones, Inc. was the ability to continue advocating for patients and helping them navigate the healthcare system.

One fun fact about me is that I spent the past year volunteering with the Jesuit Volunteer Corps. I lived with six other housemates, and we worked at different non-profits throughout Cleveland. I worked with refugees and immigrants at a federally qualified healthcare center, where I helped to reduce barriers to care and promote resources and community events.

This experience allowed me to recognize and address gaps in our healthcare system. I am eager to dive deeper into the scientific aspects of rare diseases like HPP and put my undergraduate studies to use. I enjoy listening to people's stories and comprehending research that enhances our understanding of care and treatment options. I am excited to connect with the HPP community and contribute to Soft Bones.

## Meet Lucy George

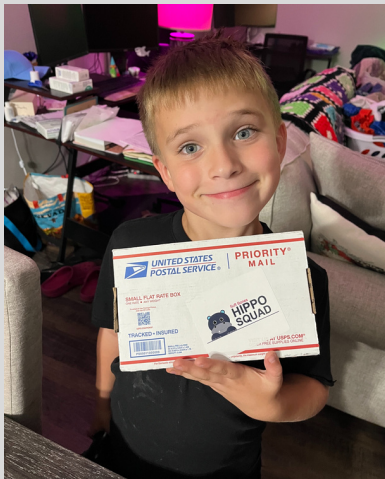
Hi! My name is Lucy George. I joined Soft Bones, Inc. as the Patient Engagement Coordinator and Administrative Assistant in November. I love talking to patients and helping them in any way I can. I interact with patients, aligning them with educational materials and program offerings. I am thankful to be a part of such a great team that supports HPP patients.

A fun fact about me is that I am the youngest of 6 kids! Growing up with such a big family taught me teamwork, patience, and how to be heard. I believe these skills help me with my work every day! I'm a wonderful listener with a passion for helping people. I am so happy to be a part of this team! If you ever need anything, please reach out, and I will be the friendly voice on the other end!

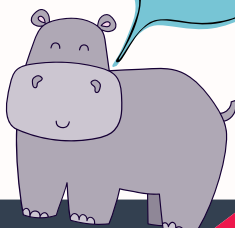


**PATIENT/CAREGIVER  
ENGAGEMENT  
COORDINATOR AND  
ADMINISTRATIVE  
ASSISTANT**

# Program Updates



JOIN THE  
HIPPO  
SQUAD



## Hippo Squad

Calling all kids ages 3-12! Have you joined the Soft Bones Hippo Squad? Hippo Squad enrollees receive a box full of fun and educational activities each quarter. This program is open to kids with HPP, their siblings, and children of parent(s) with HPP. Currently, there are 116 families enrolled in the program, with 121 children receiving the Hippo Squad mailings. This program is open to US residents only. Sign up [here](#).

The theme of the fall box emphasized that when people come together, they can make an impact. Just like a band ensemble that combines individual talents to make beautiful music, we can share our stories and experiences about HPP and help teach others about HPP. When we combine our stories, we can make a real difference. Each squad member received a variety of instruments and coloring pages.

## Stick it to HPP

The Soft Bones "Stick it to HPP" program developed by our Teen Advisory Council (TAC) aims to aid children and teens who are ready to start administering their injections. There are currently nine participants enrolled in the program.

Upon signing up, participants receive a starter kit with educational and motivational materials, including injection goals, achievements acknowledgment, tips for reducing needle and injection anxiety, a stress ball, a shot blocker, an ice pack, breathing techniques, and a playlist to get excited about self-injections. Participants can also request encouraging videos or calls from other teens for support and guidance. By taking control of their health through self-injecting, kids gain more confidence and freedom to travel and partake in activities like camping and sleeping over at friends' houses. We are super proud of all of our enrollees! Register for "Stick it to HPP" [here](#).



# Patients of the Month



**Connor Eli H.**

Learn how Connor and his family navigate the intricate journey toward the correct diagnosis and treatment after breathing problems and frequent ER visits left them puzzled for months.

Read [here!](#)



**Judith H.**

Explore Judith's story, demonstrating the importance of early diagnosis and the importance of research to empower individuals with HPP to advocate for themselves and raise awareness of diverse experiences.

Read [here!](#)



**Cannon S.**

Find out why Cannon ran away from his physical therapist (PT) and how his independence has grown over the years.

Read [here!](#)

Soft & Bones  
 YOU CAN MAKE A DIFFERENCE IN THE LIFE OF AN HPP PATIENT  
 DONATE

**Watch Our Video Featuring our 2023 Patients of the Month!**



# HPP Community: Region Round Ups

## NORTHEAST REGION

On September 23, 2023, several members from the Northeast region met for coffee and tea after an Alexion Unmask HPP event in Stamford, CT. They were joined by OneSource Case Manager Diana Miller and Kara S. (an HPP star who is a caregiver to her son). For several members of the group, this was the first time they had the opportunity to talk to others with HPP (other than family members with HPP). It was great to share stories about our individual HPP journeys and feel validation for many of the issues we have experienced. Time went too quickly. Co-region lead, Judy Harris, would love to hold another meet-up soon!



## MIDWEST 1 REGION

Region Lead Cindy Reasor hosted a luncheon meeting in Scottsburg, IN, on October 21, 2023. OneSource Case Manager, Sandy Tefft, was in attendance. During lunch, participants engaged in lively conversation before participating in the Cannonball 5K. It was a beautiful and scenic day for a 5K.



## MIDWEST 2 REGION

Region Lead Cindy Reasor hosted a meet-up for coffee and conversation in Cincinnati, OH, following an Alexion Unmask HPP event on September 30, 2023. Joined by OneSource Case Manager Liz Kearny and Matt Hay, Alexion, AstraZeneca Rare Disease, they shared conversations and had a wonderful time getting to know one another.



## SOUTHWEST REGION

On October 13, 2023, the Southwest Region had a Zoom group meeting for all the HPP patients and family members. We had a lovely 2 ½ hour open chat where we talked about HPP and some fun facts about all of us. We had a great time laughing and learning about HPP from each other and our different journeys. It was great to see old friends and new members. It felt like we were all family. Amy Britt and Sue Krug plan on having another Zoom meeting in February where they will discuss plans for an in-person meet-up in the Phoenix area. We hope to see all our Utah, Arizona, Nevada, New Mexico, and So. California members on our next virtual meeting.







## Bonfire T-shirt Fundraiser for World HPP

On World HPP Day, we partnered with Bonfire Shirts, where a percentage of sales was allocated to Soft Bones. We are excited to share that the fundraiser was a resounding success, with 89 shirts sold! Thank you to Sue and Cindy for hosting such a great event!

## Lynch Creek Farm

This holiday season, we partnered with Lynch Creek Farm for a wreath fundraiser where 15% of the proceeds went to Soft Bones. Lynch Creek Farm handcrafts traditional and decorated Christmas wreaths, centerpieces, and other holiday gifts. We had at least three wreaths purchased. Thank you to those who purchased these wreaths this year. We hope you loved them!



## Bundt Cake Fundraiser

Cassandra Self, a Southwest Region Lead, hosted a local fundraiser in Texas with Nothing Bundt Cakes in October to coincide with World HPP Day. Thank you, Cassandra, for all your efforts!



# Fundraising Corner

## New Fundraising Program: RaiseRight

RaiseRight is our newest fundraiser. It is a platform where you can buy gift cards to over 750+ brands, and a percentage of the proceeds goes to Soft Bones. Use our enrollment code **LCHSIABAFERN** to join and support us through every gift card purchased!

Buy gift cards  
that can  
support us!



RaiseRight

## Shop for a Cause

Empower our mission by choosing to make an impact through your purchase of goods and services. Explore our 'Shop for a Cause' page, where you will find multiple opportunities to support our cause year-round or join us in our seasonal fundraising campaigns. Shop with a purpose and support Soft Bones!

## Facebook Fundraising

We are happy to announce that our Facebook/Instagram fundraising platform is back up and running! That means you can host fundraisers on Facebook or Instagram for your birthday, awareness days, end-of-year giving, or any other time you want to show support for Soft Bones! Please consider helping us continue to deliver on our mission by hosting a fundraiser on Facebook or Instagram today! Thank you for your support.



**Bookmark our [fundraising page](#) to support Soft Bones year round!**