

# THE BARE BONES

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

## Soft Bones Hosts Second International Scientific Meeting

The Second Scientific Meeting of Soft Bones Inc., held June 2 - 4, 2023, in Bethesda, MD, USA, brought together clinicians, physician-scientists, basic science researchers, and industry representatives from across the globe to better 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.

The Scientific Planning Committee members included Michael P. Whyte, MD, Committee Chairman, Washington University School of Medicine, Kathryn Dahir, MD, Vanderbilt University Medical Center, Cheryl Rockman-Greenberg, MD, Max Rady College of Medicine University of Manitoba, José Luis Millán, PhD, Sanford Children's Health Research Center, and Peter Tebben, MD, Mayo Clinic.

This Second Scientific meeting was organized by Deborah Fowler, President and Founder of the Soft Bones Foundation, and assisted by Denise Goodbar, LeighAnne Castimore, Katie Kelly, Sue Krug, Ellen Reppe, Cindy Reasor, and consultant, Charlene Waldman, along with an on-site technical team. Special thanks to the sponsors: Alexion AstraZeneca Rare Disease, AM Pharma, Charles River, PuREC, Rallybio, Rampart Bio, Be Biopharma, and IcBio.



The full agenda of the meeting can be found [here](#).

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**An article including the abstracts presented at the meeting is available on JBMR Plus® [here](#). On-demand videos will be available for clinicians and researchers on HPP Connect soon.**

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# UPCOMING EVENTS

## Annual Golf Classic: Register Now!



Registration is now open for our largest fundraiser of the year! The 15th Annual Soft Bones Golf Classic will be held on Monday October 2, 2023 at Somerset Hills Country Club in Bernardsville, NJ.

Click [here](#) to register and view sponsorship opportunities.

If you are interested in volunteering or donating an item for the raffle, please reach out to [info@softbones.org](mailto:info@softbones.org). Thank you to our tournament sponsor, Atlantic Health System.

## Join us for our Second Annual Cannonball Run, Roll, Stroll

This year, in honor of World HPP Day, Soft Bones will once again host the Cannonball 5k Run, Stroll or Roll, a virtual run/walk. Anyone interested in participating is to complete the registration, print a race bib and complete a 5k run/walk at your convenience, anytime, anywhere between October 1-30, 2023.

Register for the Cannonball 5k Run, Stroll or Roll [here](#).

World HPP Day is our chance to spread awareness about hypophosphatasia and bring the entire HPP community together, including patients living with HPP, caregivers, families, loved ones, researchers and doctors. There's an opportunity for everyone to get involved!

To purchase Cannonball 5k and World HPP Day apparel, visit our [shop here](#).



Thank you to Alexion AstraZeneca Rare Disease for their support of this event and of our HPP Community!

# Events Recap

## Denim & Diamonds

Our first-ever Denim and Diamonds fundraiser was a resounding success! With more than 60 people registered, the rain cleared way for a beautiful evening and attendees danced the night away alongside horse pastures and cornfields. Here are a few photos from the event.

A special thank you to our sponsors: Green Room Communications, Equity Wine Ventures, UEM Events and Design, DS Cake Designs, Hatfield Schwartz Law Group, Catz 'n Dawgz, and Boutique Calia. Also thank you to everyone who donated auction items and helped to make the event a success.



## Fun at Trivia Night



Congratulations to the TAC team members who hosted the successful first-ever Trivia Night on May 23rd. Members of the HPP community along with friends and family came together to test their knowledge, compete for prizes, and have an all around terrific time. Though joining the event was free, the TAC raised almost \$600 through donations!

Keep an eye out for more TAC sponsored events coming soon!

# Stick It to HPP

The Teen Advisory Council (TAC) recently launched "**Stick It to HPP**", a program aimed at aiding children and teens who are ready to start administering their own injections. The TAC is excited to announce that there are currently six patients 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 partake in activities like camp, traveling, and sleeping over at friends' houses. We are super proud of all of our enrollees!

Register for "**Stick it to HPP**" [here](#).

## HIPPO SQUAD

**Calling all kids ages 3-12!** Have you joined the Soft Bones Hippo Squad? Hippo Squad enrollees receive boxes full of educational and fun activities each quarter. This program is open to kids with HPP and their siblings as well as children of parent(s) with HPP. There are currently 96 families enrolled in the program with 121 children receiving the Hippo Squad mailings. This program is open to US residents only. Enroll [here](#).

The theme of the summer box emphasized the importance of educating others about HPP. Kids received everything needed to create their own puppets along with a script that they were encouraged to use to learn how to talk about HPP.



# Patient of the Month

Every month, Soft Bones highlights a member of the HPP community to share their personal diagnosis experience and journey. If you feel your story could help another person not feel alone, be sure to reach out to us and share your experience. Since our Spring Newsletter, six new courageous hypophosphatasia patients have been featured as Patients of the Month!

## MEET HALEY G.

For the month of April, **Haley G.** highlighted her remarkable HPP journey. Haley, who was diagnosed at birth but is now in her thirties, shared how her symptoms really started to present themselves in her mid-twenties. This made it hard for her to work full-time, go to school, and have a social life. By 2019, Haley started taking enzyme replacement therapy and has experienced a great deal of relief and success from her injections. She also credits a lot of her success to the HPP community she has found in Soft Bones, meeting other patients and finding life-long connections and friendships with people in similar situations to hers!



MAY



## MEET EVAN W.

In May, **Evan W.**, a baby boy of loving and supportive parents, was featured as Patient of the Month. Before he was born, his parents were aware that something about Evan's growth progress in the womb was alarming, and his doctor's suspected HPP. After multiple tests, failed extubation attempts, and surgeries, Evan was officially diagnosed with hypophosphatasia at one month old. Since then, he has progressed immensely! Though it has not been all smooth sailing, Evan's mom, Kodi, says that he smiles every chance he gets and the support and knowledge from the Soft Bones community has helped get them through this difficult journey for their baby boy.

# Patient of the Month

## MEET AUGUST B.

In June, another amazing baby boy, **August B.**, was featured as Patient of the Month. With his limbs progressing abnormally before he was even born, an amniocentesis was performed confirming that August did indeed have HPP. From the minute he was born, he was put under immediate observation, managing to show his new, beautiful personality through every part of the journey. Currently, August no longer has to be on a ventilator or a g-tube. He has learned a great deal of sign language since he lost some of his hearing along his journey, and enjoys watching Monsters Inc everyday!



JUNE

JULY



## MEET NATALIE S.

For the month of July, **Natalie S.** was spotlighted. This brave four-year-old girl has been on her HPP journey since she was diagnosed at just three days old. Thankfully, she was put on enzyme replacement therapy almost immediately following, and at six-months-old she found her forever home with her adopted family. After doctors noted increased pressure on her optic nerve, Natalie was also diagnosed with craniosynostosis, or premature fusion of the skull, which is not uncommon in HPP. After surgery and five months with an eye patch, her distractors were removed and she has been hospital free since! She strives to bring hope to other kids struggling with their HPP journeys through her own triumph!

## MEET MASON P.

In August, **Mason P.** was featured as Soft Bones' Patient of the Month. At three years old, family and friends were commenting on the way he ran, and his parents looked into it further. They noticed that he lacked a bend in his knees when jumping and running and brought it up at his check up appointment with his doctor. From the x-rays, it was visible that he showed bone demineralization in his legs, and his lab work revealed low ALP levels. He also started to lose his teeth prematurely at the age of two, a telltale sign of HPP. He was soon diagnosed and put on enzyme replacement therapy, just one month after diagnosis. Since then, Mason improved so much! He is also a member of Soft Bones Hippo Squad and he and his family are so thankful for all of the knowledge and support they have been given from Soft Bones on his journey!



SEPT



## MEET PEYTON W.

In August, 10-year-old **Peyton W.** was spotlighted as the Patient of the Month. She was diagnosed with juvenile onset HPP in December 2022, just 9 months ago, after a series of unusual fractures and injuries. Peyton's journey with HPP began in November 2021 when she fractured her first bone, followed by complications such as Complex Regional Pain Syndrome and a hip injury. Her unusual injuries led her doctor to order lab tests, revealing low ALP levels and high B6 levels. Shortly after, Peyton was officially diagnosed with HPP. Peyton and her family have found support from Soft Bones and Alexion. Peyton has improved tremendously, and now self-administers Strensiq injections. Peyton offers encouragement to others facing similar challenges, emphasizing that the injections become more manageable with time and perseverance.

**READ THEIR FULL STORIES AND LEARN MORE ON  
OUR WEBSITE**

## Employee Spotlight - Cindy Reasor: The Project Coordinator Drawing on Personal Experience to Help Others



Cindy Reasor joined Soft Bones as the Project Coordinator in October of 2021. She is thrilled to be putting her life experience with HPP into practical use at Soft Bones. Cindy has a passion for helping others navigate life with a rare disease, advocating and taking a proactive part of the organization's mission. She is grateful to be a part of such a great team that supports HPP patients daily. Cindy interacts with patients, aligning them with educational materials and program offerings. She also serves as our Events Coordinator for our Regional and National meetings, manages our community support website, HPP AND ME, as well as moderates our social media channels.

Cindy is married with three grown children and six grandchildren. A number of Cindy's family members are also affected by HPP. Her two daughters are both nurses and went into the healthcare field to help add compassion and understanding care where it is much needed. Cindy currently resides in Southern Indiana and also serves as a fundraising volunteer and a Region Lead for the Midwest 1 Region.

## Soft Bones Grant Winner Dr. Dobrawa Napierala Receives 2023 Adele L. Boskey Award

***Congratulations to Dobrawa Napierala, Ph.D. for receiving the 2023 Adele L. Boskey Award! This award will be presented during the ASBMR 2023 Annual Meeting at the Vancouver Convention Centre in Vancouver, BC, Canada.***

Dr. Dobrawa Napierala, Ph.D. is an associate professor in the Department of Oral and Craniofacial Sciences, University of Pittsburgh School of Dental Medicine. She studies molecular and cellular mechanisms regulating physiological mineralization, and mineralization pathologies in human genetic diseases. Dr. Napierala is a dedicated mentor supporting a diverse group of trainees and junior faculty. She is a director of the Oral and Craniofacial Sciences graduate program and an assistant director on the NIH/NIDCR-funded Pittsburgh Craniofacial Sciences Training Program. Dr. Napierala serves the research community as a co-director of the Pittsburgh Center for Interdisciplinary Bone and Mineral Research, member of the ASBMR Innovation Committee and a member of the Board of Directors of the International Conferences on the Chemistry and Biology of Mineralized Tissues.





# Upcoming Opportunities

## Research Grant Applications Open

The Soft Bones Foundation is offering a "seed grant" of \$25,000 for research in 2023-2024 that is directly related to hypophosphatasia. This will be the 11th grant awarded by Soft Bones since 2014. Proposals are welcome from faculty members at a college or university or investigators at private research institutions.

Applications from senior postdoctoral trainees or research associates are acceptable provided that their mentor has a record of commitment to research relevant to HPP. **Click [here](#) to learn more.**

## SAVE THE DATE

### Soft Bones 2024 National Patient Meeting

July 12-13th, 2024

Great Wolf Lodge

Mason, OH

Registration Information Coming Soon!

## HPP AND ME Community Zoom Chat

Join us for an HPP AND ME Community Chat on **September 19, 2023 at 1pm EDT**. No registration required, just click the link to join: <https://us06web.zoom.us/j/88116274370>  
Community chats are a great, informal way to get to know the faces behind the screen names. The conversations range from HPP to our pets and travels.



With so many concerns about patient privacy online, Soft Bones created the online community HPP AND ME for people living with HPP. By becoming a part of the HPP AND ME community, you can inquire about concerns, receive support, exchange your experiences, and keep up with the most recent research, significant news, and announcements. Membership to the website is free and is growing everyday! The site is organized by topic and can be easily searched. A diverse range of discussions are taking place, such as seeking a diagnosis, life after diagnosis, and more. You can also find introductions and backgrounds of patients on their journeys so far.

*If you have any questions or need assistance, please email [Cindy@SoftBones.org](mailto:Cindy@SoftBones.org)*

# Remembering Gerald Brandt

It is with shock and great sadness that we announce the passing of Gerald Brandt on May 19, 2023. Many of you have come to know Gerald as he was an outspoken advocate for the hypophosphatasia community and the founder of HPP Deutschland eV, one of the first HPP patient advocacy groups ever created. However, some of you may not be aware of his work and impact on the HPP community, including the formation of Soft Bones.



As an accomplished writer in English and German, Gerald shared his lifelong stories of living with the disease. He learned to walk

late, described himself as not very resilient as a child, endured early tooth loss, and developed strong knock knees. After a number of operations and numerous fractures during childhood and into adulthood, he then endured joint problems and pronounced kidney dysfunction. According to his own statements, he stopped counting the number of broken bones.

He founded HPP Deutschland eV in 2006 to bring together the HPP community in Germany, hosting annual patient meetings, advocating with policymakers for HPP to be recognized as a disease, and spending countless hours on the phone with patients and families, supporting and educating as well as connecting them with an appropriate care team. He was also sought after as a speaker and published author, hypothesizing that King Tutankhamun had hypophosphatasia. <https://pubmed.ncbi.nlm.nih.gov/24466636/> Gerald had a tender heart for animals and a great love of music. In recent months, he spent his time restoring guitars and donating them to local children in need. He was trained in guitar and recently started learning the ukulele.

Many of us will remember Gerald for being a lifeline in times of need. He supported patients and families from around the world, helping to answer questions and continuing to raise awareness of HPP. He has been an incredible resource to many, and we are forever grateful for his time, dedication, support, and friendship. He is greatly missed by all of us at Soft Bones and the impact of his work will continue for years to come. As the German HPP group shared, "You can close your eyes and wish for him to come back or you can open them and see what he left behind."

From what we understand, Gerald passed during an emergency medical procedure that was unrelated to HPP. This is all the information we have at this time. We are respecting the family's wish to refrain from further inquiries at this time.

We are so sorry to be the bearers of this sad news.

# Remembering Dr. Philippe Crine



2023 has been a difficult time of loss in our HPP community. We are saddened to announce the passing of Dr. Philippe Crine.

Many of you may have never heard of Dr. Crine. He's the scientist who discovered the first and only approved treatment for HPP, asfotase alfa, and was the founder of Enobia Pharmaceuticals. Many of our patients had the privilege of meeting him face-to-face at past Advocacy in Action meetings. Please keep his family in your thoughts and prayers during this difficult time.

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Philippe Crine passed away on June 13, 2023. He was swept away by pancreatic cancer, within the span of two months. He died peacefully, at home, surrounded by his loved ones.

A Professor of Biochemistry at the Université de Montréal, he founded Enobia Pharma in Montreal in 1997. Enobia Pharma was a start up in biotechnology that developed a specific expertise in bone related orphan diseases. The company developed a bone targeted enzyme replacement therapy for hypophosphatasia, a rare and often fatal metabolic bone inherited disease. There was no approved therapy for hypophosphatasia at the time.

Philippe will be remembered as the man who, with a team of dedicated and passionate researchers, gave a new lease on life to children born with hypophosphatasia. The innovative therapy allowed children with the disease not only to survive but to lead normal lives. Forever humble, he considered himself and his team privileged to have made such a huge difference in the lives of so many children and families.

His visions and his seminal contributions to the study of bone structure and rare diseases impacted a whole generation of students, researchers, and collaborators. Until the very end, he was passionate about sharing his knowledge and expertise. One month before his passing, he gave a keynote address at a colloquium aimed at young researchers interested in starting their own companies. He was adamant that he should share the ups and downs of Enobia Pharma with his audience of young scientists. Even his growing cancer would not stop him from participating in the conference. His zest for life was phenomenal.

# HPP Community: Region Roundup

## MIDWEST 1 REGION

The first in-person region meeting in Minnesota was hosted on August 13th by Region Lead, Nicole Belting. Thirteen people from across the region attended as well as OneSource Case Manager, Krista Higgins. Lunch was served and the group sat around Nicole’s dining room table and had conversations about HPP. A good time was had by all!



## SOUTH CENTRAL REGION

On August 17th, the South Central Region Co-Lead, Cassandra Self, hosted a casual lunch in Dallas for patients and caregivers in the area. Deborah Fowler was also on hand for the meet-up. The group discussed plans for the upcoming Cannonball Run, Roll or Stroll as well as ways to bolster HPP education of local doctors in the Dallas/Fort Worth area.



## MIDWEST 2 REGION

Midwest 2 Region Co-Lead, Sharon Talkington, gathered with some members of the region for lunch and an afternoon of conversation on August 26th in Ohio.

The Midwest 2 Region is comprised of members from Kentucky, Michigan, Ohio, West Virginia.



**Not sure what region you’re in, or who to connect to?  
Find out using the [Soft Bones region map](#).**

# Fundraising Corner



**Sending out high-fives to our community members who recently hosted their own fundraising events for Soft Bones:**

*Sharon Talkington for hosting a Mary Kay Fundraiser*

*Blynda Kellner for hosting a Chick-Fil-A Fundraiser*



**THANK  
you**

If you have an idea for your own fundraiser or want support from our team, contact [info@softbones.org](mailto:info@softbones.org). We are happy to help!

## Soft Bones 2022 Annual Report

The Soft Bones 2022 Annual Report is available on our [website](#). Read about some of our exciting accomplishments from 2022.