

# Soft Bones

## 2022 Annual Report



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Dear HPP Friends:

2022 was quite a whirlwind for us at Soft Bones.

We finished the year on a high note, conducting our own Externally-Led Patient-Focused Drug Development meeting, also known as an EL-PFDD. The EL-PFDD gives a voice to patients, allowing them to share their experiences living with hypophosphatasia, including their successes and challenges with therapy, impact on their quality of life, and hopes for future treatments. We were thrilled to have several members of the US Food and Drug Administration attend, along with many physicians, healthcare practitioners and of course, patients and families from around the world. You can read more about our EL-PFDD [here](#).

Another big accomplishment was that we emerged from the COVID-19 pandemic and once again were able to gather in person, coming together for the 2022 National Patient Meeting in Florida. However, one lesson learned from the pandemic was that virtual meetings are more greatly accepted and appreciated. We always want to remain conscious and inclusive of everyone, which is why we continue to invest in hosting hybrid meetings where attendees will have the option to be present in person or online. Our Soft Bones team understands that not everyone is able to attend in-person meetings, and we want to make sure that no one misses out.

As we navigate the ever-changing challenges of today, we continue to grow our educational programs, along with holding several successful events, meetings and fundraisers. Alongside the National Meeting, we held a Southeast Region Meeting, South Central Region Meeting, a Southwest Region Meeting and attended the American Society for Bone and Mineral Research (ASBMR) Annual Meeting, and many more. Our 2022 World HPP Day theme, "HPP On the Move" celebrated the importance of physical activity along with the movement to raise awareness for HPP. We're also very excited to announce the launching of a new HPP symptom tracker to help patients and caregivers more easily capture important details and empower them to have more productive conversations with their doctors. Keep an eye out for more information on this exciting initiative.

We are beyond appreciative of everyone who gave their energy and attention to assisting our organization achieve its goals this past year. Thank you to each of you who contributed to our foundation by donating what you could. And of course, thank you always to our amazing Region Leads who do such an incredible job volunteering their time watching and listening at the community level in order to support patients' needs and ensure everyone is being represented and heard.

*Deborah Fowler*

President and Founder  
Soft Bones

# Board of Directors



**Deborah Nettune Fowler**  
President and Founder



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Attorney



**Rob Moskow, Esq.**  
Attorney



**Scott Nettune**  
Board Member



**Brenda Ziegler, DO**  
Board Member

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The last two years have been difficult, but our supporters have stepped up to help us reach our fundraising goals. As a result, we have become a stronger organization bolstering awareness of HPP and have a renewed focus on supporting patient access to healthcare providers who have experience with this disease and to appropriate treatments.

## Staff

**Deborah Fowler**, President and Founder  
**Denise Goodbar**, Executive Director  
**Sue Krug**, Patient Liaison  
**Cindy Reasor**, Project Coordinator  
**Katie Kelly**, Database/CRM and Event Coordinator

Special thanks to our amazing staff who work tirelessly to keep Soft Bones running, stronger than ever before!



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## Our Mission

The mission of Soft Bones, Inc. is to provide valuable information, education and support for people living with hypophosphatasia (HPP), their families and caregivers. The Foundation promotes research of this rare bone disease through awareness and fundraising efforts.

## About Soft Bones

Soft Bones is a 501(c)3 non-profit, US-based patient advocacy group dedicated to the community of patients, caregivers and families living with hypophosphatasia. Soft Bones powers the HPP community by raising awareness of this rare, metabolic disease, providing hope by advocating for and funding research, and connecting patients with each other and with the medical community to advance the understanding of HPP. We provide medical information as well as a support system for patients and families living with HPP.

We believe a cure is possible. With modern-day science of gene editing and gene therapy, we work closely with researchers and clinicians to champion collaboration, reduce duplication of efforts, share findings and unite the global HPP community. We also work to position HPP as a favorable subject of research.

Hypophosphatasia is a serious condition. Scientists are still working to better understand the impact of low levels of alkaline phosphatase in the body. While the hallmark of HPP is soft bones, as a metabolic disease, the full impact is not yet fully understood.

## Scientific Advisory Board

**Michael Whyte, MD, Chair**  
Shriners Hospitals For Children

**Thomas Carpenter, MD**  
Yale School of Medicine

**Kathryn McCrystal Dahir, MD**  
Vanderbilt University Medical Center

**Matthew Drake, MD, PhD**  
Mayo Clinic

**Gary Gottesman, MD**  
Washington University  
School of Medicine

**José Luis Millan, PhD**  
Sanford Children's Health  
Research Center

**Lothar Seefried, MD**  
University of Würzburg, Germany

**Jill Simmons, MD, DDS**  
Vanderbilt University Medical Center

**Peter Tebben, MD**  
Mayo Clinic

**Timothy Wright, MS, DDS**  
University of North Carolina Chapel  
Hill, School of Dentistry

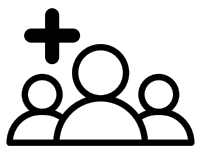
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## Region Leads

**Chris Denune**, Northeast  
**Judith Harris**, Northeast  
**Blynda Kellner**, Southeast  
**Kirsten Mitchell**, Southeast  
**Cindy Reasor**, Midwest I  
**Nicole Belting**, Midwest 1  
**Nichole Macknight**, Midwest II  
**Sharon Talkington**, Midwest II  
**Susan Brown**, Central  
**Lindsey Willson**, Central  
**Nellie Sanders**, Central  
**Kara Schweiss**, Central  
**Cami Rush**, South Central  
**Cassandra Self**, South Central  
**Amy Britt**, Southwest  
**Sue Krug**, Southwest  
**Ann Haak**, Northwest



# Our Year in Review



**216+** new patients this year

**220+** information packets sent to newly diagnosed or suspecting HPP individuals\*



Community members in **37** countries plus Puerto Rico

**437** known Mutations



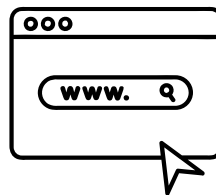
Over **\$25k** awarded in grants\*\*

HPP TeleECHO Sessions  
Jan 1 2022 - Dec 2022  
Nearly **800** total registrants



## 2023 Website Stats

**40,995** total users  
**179,929** page views

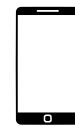


**5** Podcast Episodes Recorded  
**100+** Downloads



Most Visited Page  
Low ALP, Could it be HPP?

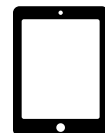
How Visitors View It



Mobile  
**59.95%**



Desktop  
**37.66%**



Tablet  
**2.17%**

## Top 10 Countries

**1. US - 81%**

Top States NY and CA

**2. India**

**3. Canada**

**4. UK**

**5. Australia**

**6. New Zealand**

**7. Japan**

**8. Philippines**

**9. Germany**

**10. China**

\* 70% requested mailed packets; 30% requested emails

\*\* Soft Bones has awarded over \$475k in grants since its inception.

# Rare Disease Day



## Rare Disease Day Webinar

In partnership with Aruvant Sciences, Soft Bones hosted a gene therapy webinar to discuss and educate its promises and challenges for hypophosphatasia. Special thanks to our presenters, Dr. Jose Luis Millan from Sanford Burnham Prebys and Matt Grol of Western University. The webinar lives on the Soft Bones website for future reference. [Click here](#) to view the gene therapy webinar.

## RDD POLL

Our 2022 Rare Disease Day campaign also included a poll of patients to highlight some challenges of living with HPP. We encouraged participants to share these results with their network and help bring awareness to the struggles faced.

# Rare Disease Day

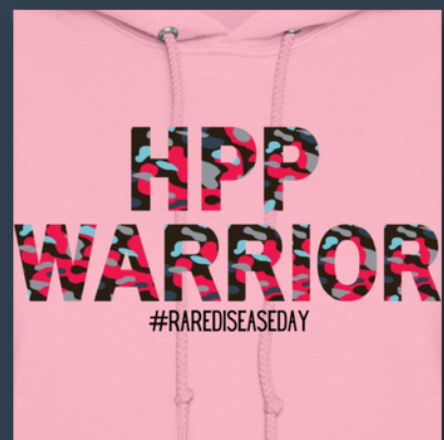
## Coloring Contest

To bring awareness to the wide range of symptoms that affect patients living with HPP, adults and children alike completed a coloring activity sheet and shared it with their friends and family on social media. All who participated were entered to win a Soft Bones hippo!



## HPP Warrior Apparel

A new line of “HPP Warrior” apparel launched on our website store. The apparel never goes out of style. View the designs [here](#).



# We Are Stronger Together: Raising Our Voices for HPP

## Celebrating World HPP Day with *Movement*

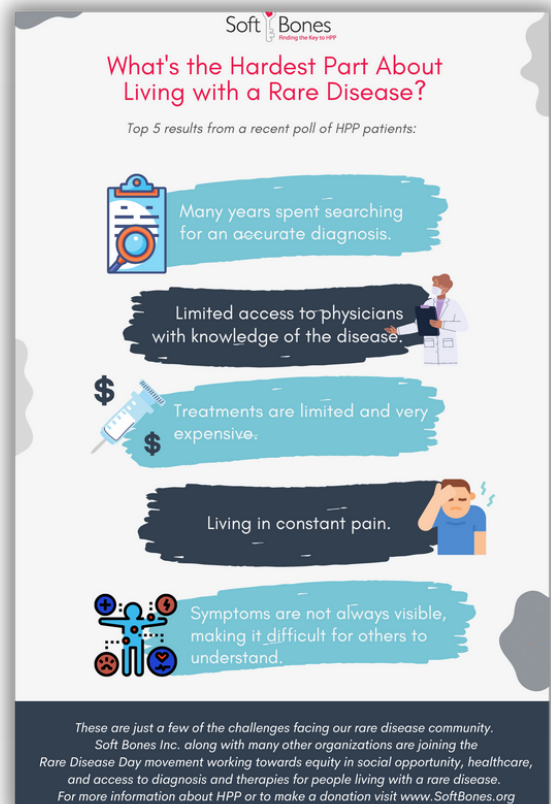
Each year, we celebrate World HPP Day on October 30th. Our 2022 theme “**HPP ON THE MOVE**” was a celebration of movement both physically and metaphorically. Whether people chose to get moving by participating in the Cannonball 5k or another form of physical activity – or started their own movement by educating others – the goal was to create momentum around awareness of hypophosphatasia around the world. *Every October, we look forward to this special day to celebrate how far we’ve come in expanding awareness about HPP.*

## Strengthening Support for Rare Disease Day

Rare Disease Day provides an energy and focal point that enables rare disease advocacy work to progress locally, nationally, and internationally. For HPP, it allows individuals to share stories and come together with the global rare disease community to raise awareness for HPP and the needs of our patients. On February 28th, 2022 Soft Bones joined many other organizations to commemorate the day and show support for those living with diseases that may fly under the radar.

## Taking Care of HPP Patients: Launching the “Take Care” Program

Early in 2022, we launched our “Take Care” program. The kits include a variety of age appropriate, activities and items to infuse a bit of fun into the day. The contents of each kit are tailored to the unique needs of the patient receiving it, which gives it an extra special meaning to them. Kits are meant to serve as a pick-me-up for patients who may not be feeling their best, letting them know someone is there to Take Care of them when they need it most.



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# There is **Power in Education**

## National Patient Meeting In-Person and On-Demand

On July 9th, the 2022 National Patient Meeting was held in Orlando, Florida. The meeting gathered 150 attendees, patients, caregivers and exhibitors, making it our first in-person gathering since the pandemic! Special thanks to our sponsors, Alexion AstraZeneca Rare Disease, PANTHERx Rare and Aruvant Sciences who made it possible for us to host this meeting that included an art workshop, where patients were guided through a session to illustrate their HPP journeys on canvas with watercolor. For those who were unable to attend, all sessions are available [HERE](#) on our virtual platform. 2023's National Patient Meeting was held February 17-18th in Phoenix, Arizona!





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# American Society for Bone and Mineral Research Annual Meeting



This year's ASBMR 2022 Annual Meeting in Austin, Texas was a huge success! With more than 2,000 attendees, 100 education sessions, 1,000+ poster presentations, and an exhibit hall with many booths, we had endless opportunities to catch up with healthcare practitioners, and researchers with an interest in HPP.

Our Soft Bones team connected with doctors from all over the world interested in learning about HPP. We also were able to collaborate with new researchers to further help patients with rare bone disease complications.

ASBMR featured presentations from multiple speakers, such as Dr. Michael Whyte who discussed a new Vitamin B6 study, and Dr. Gaddy, the winner of the 2019 [Soft Bones Research Grant](#), who discussed the team's research into the effects of maternal HPP on development in sheep in utero.

When our teams are able to attend more conferences and meetings, it allows us to expand and learn more about how we can help those impacted by HPP.

*The latest results were presented at a Soft Bones TeleECHO, and physicians and researchers can now access it on HPP Connect, an online community created for physicians and healthcare professionals to learn more about HPP.*

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## Regional Reach: Touching Lives One Community at a Time

Soft Bones is focused on supporting HPP patients at the community level. We have Region Leads who continuously build relationships with patients in their respective geographical areas with the goal of hosting in-person, community-based meetings, to help provide patients with the care and education they need. From small towns to big cities, Soft Bones is there every step of the way.

### South Central Region

During the American Society for Bone and Mineral Research Annual Meeting (ASBMR), we were thrilled to have the opportunity to host a luncheon for the South Central Region in Austin, Texas. The intimate group meeting was powerfully productive, allowing us to gain so much over delicious tacos! Dr. Gottesman joined us and we discussed the difficulty carriers of HPP face in gaining a diagnosis and treatment plan for HPP.



Dr. Gottesman shared a wealth of knowledge and new tests that can be requested to help determine if a HPP diagnosis is the right path or other treatment can be used. These meetings allow us to gain valuable insights into the expertise of our valuable doctors. If you're interested in gathering at one of our region meetings, please email [info@softbones.org](mailto:info@softbones.org).

### Bō-nə-fīde HPP Podcast



Soft Bones podcast, Bō-nə-fīde HPP, is designed to educate and support the families and caregivers of those affected by HPP. As a mother and caregiver, host Deborah Fowler and her guests discuss this rare genetic bone disease with people from all over the globe. Bō-nə-fīde HPP can be found on [Spotify](#) or in the [Soft Bones Resource Library](#) on our website. In 2022, five episodes were recorded and the podcast was downloaded more than 100 times!

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## Externally-Led Patient-Focused Drug Development (EL-PFDD)

This year, Soft Bones launched the first Externally-Led Patient-Focused Drug Development (EL-PFDD) meeting on November 15th, hosted by Deborah Fowler, President and Founder of Soft Bones, and James Valentine, meeting moderator. Hundreds of individuals from the HPP community listened, called and wrote in to show their support and to participate in spreading awareness for HPP. The EL-PFDD was an incredible opportunity for representation for the HPP community, as the FDA heard from a remarkably diverse group of patients, caregivers, and experts. Individuals of our community took part by sharing their stories, serving as panelists, and voting in the live polling sessions held during the meeting.



## Strength in Numbers: Two Sisters Living with HPP

Amy and Suzanne are adult twin sisters who have been living with symptoms of HPP for their entire lives. Difficulties began as early as infancy when Amy spent over a month in the hospital. Growing up they knew that something was not quite right. They struggled to keep up with other kids at school and Amy suffered emotionally with a speech problem for years.

Suzanne's first memory of doctors is her mother telling her not to tell the doctor about her headaches. Her mother's life experience was that doctors would not be able to find anything wrong and dismiss her symptoms. As she got older, Suzanne's symptoms continued to grow, including: loose teeth, brain fog, depression, anxiety, bone pain, unexplained joint problems, migraines, muscle aches, eye problems, and ringing in the ears. Her experience with doctors began to mirror her mother's. Doctor after doctor told her that her symptoms could be resolved by losing weight, eating better, and sleeping more.



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Amy and Suzanne seemed to be on an endless search for ways to alleviate their symptoms and pain. After talking with their female family members across generations they realized that many of them had odd overlapping symptoms. One of Amy and Suzanne's nieces led the way doing her own research and discovered she had dysautonomia. Others in the family were diagnosed with the same, but this still did not explain all of their symptoms.

Amy decided to order genetic testing privately when doctors would not order it for her. Her test results came back with a disease she had not heard of before, hypophosphatasia. Suzanne was not so sure that this was what they had based on their initial research and the symptoms described.



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**It's hard to imagine our lives without Soft Bones, mostly because the reality is that it would be so sad.**

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Amy then found the Soft Bones Facebook group. When Suzanne joined the Soft Bones group on Facebook she saw herself and her lifetime of symptoms in the posts and comments of others there. There was nowhere online that talked about the wide variety of symptoms as well as the members of the Soft Bones group.

Understanding the experiences of so many others with HPP convinced Suzanne that they belonged in the group. After receiving genetic confirmation, Amy and Suzanne went to a geneticist said to specialize in HPP. They were surprised when they were told that they did not have HPP and the doctor had no answers for them regarding their genetic mutation. They were crushed. That's when they reached out to Soft Bones and spoke with Deborah and Denise.

“Thinking about that conversation brings back tears. They believed us. They gave us the names of the best HPP doctors in the country and a path forward. It was a life-changing conversation,” says Amy and Suzanne. “It's hard to imagine our lives without Soft Bones, mostly because the reality is that it would be so sad. Without Soft Bones, we would have had to settle for a doctor who told us to ‘stop going to doctors,’ and, ‘wait ten years for science to catch up with you,’ as our primary source of help. We don't know if we would have given up, or what that would have looked like, but we feel overwhelmingly grateful every single day. We don't know what our lives would look like now without Soft Bones.”



A close-up photograph of a baby, Liam, lying on a patterned blanket. The baby is smiling and looking towards the camera. The blanket has a light-colored background with small, colorful patterns. The baby is wearing a blue and green striped shirt.

## An Early Diagnosis: Liam's Story

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We are extremely grateful for the support and information we have received from Soft Bones.

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Breeanna, Liam's Mom

Liam Christensen was born March 4th, 2022 in Idaho. He was a full-term baby and was induced at exactly 40 weeks. Liam was born with a heart murmur and left the hospital with a mild case of jaundice, which led to frequent visits to the family pediatrician. Like most newborns, this seemed normal, until it wasn't. Once Liam cleared his initial visits, his parents noticed that he was not gaining weight. The lack of weight gain prompted more frequent visits to the pediatrician to monitor his weight. However, Liam started to refuse his feedings and began experiencing failure to thrive, admitting him to the hospital.

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There were concerns about dehydration. As a result, the hospital ran more extensive tests which uncovered his labs were off. His calcium level was at 14, so they officially hospitalized him. Liam was in the hospital for about one week, where he was hooked up to an IV and they inserted an NG feeding tube. Doctors were puzzled by the fact that he didn't seem to fit into any diagnosis, so they recommended a second opinion at Primary Children's Hospital in Utah.

The family was greeted by a team of ten people. This team was partnered with geneticists, endocrinologists, and neurologists. Over a few days of more labs and testing they had two different potential diagnoses. The endocrinology team felt confident that Liam had HPP.

One of the doctors, Dr. Al Hamad, had the opportunity to visit a few children with HPP and saw the signs to help first identify the diagnosis. Because Strensiq was a special medication, they needed specific confirmation, so while they were waiting for the genetics test (which takes about 4 weeks for results) they evaluated his B6 levels, which only took 3 days for the results. With the results of his test, the doctoral team was more confident in the HPP diagnosis.

Liam was at Primary Children's Hospital for a month before he was diagnosed. Once he was diagnosed, the team worked tirelessly on getting approval for Strensiq. After 2 long months, Liam and his family left Primary Children's Hospital and upon their arrival home, they were trained by a home healthcare nurse who showed them how to properly inject and dose over a 3-hour training.

At home, the family has multiple weekly therapies such as speech therapy, occupational therapy, physical therapy, home health, and occasional lab work. Weekly, the family tracks Liam's weight, height, and head circumference. Due to Liam's weak muscle tone in his throat, he developed an oral aversion and was unable to take large amounts of liquids by mouth, which resulted in him having a G-Tube placed at 6 months old. Liam is now 15 months old, and he receives Strensiq injections 3 times a week and is slowly working on taking food by mouth. Although he has had a challenging first several months of life, he is a happy and determined baby who loves to laugh, smile and talk to everyone he meets. He is a very curious baby and likes to explore his surroundings.

Since there are very few babies that have known HPP with such an early diagnosis and use of Strensiq, there is very little information and data to reference to determine what Liam's physical, mental, and emotional health will look like in the future. The family actively documents Liam's progress through social media in hopes to raise awareness and potentially help any future families with their own HPP journey.



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# Connecting The Dots: The Growth of Our HPP Network

## A Giant Leap for the Establishment of an HPP Network

In 2022, we took a giant leap forward as we established our growing HPP network (formerly Centers of Expertise). Through the generous contributions and dedication of our diverse steering committee and working group members, who are made up of patients, parents, nurses, geneticists, physicians, physical therapists, and more, provided input and guidance to outline a plan for the creation of a framework for comprehensive patient care and better quality of patient care. To accomplish this, we have identified five key strategies: 1) increasing expertise in HPP (e.g. virtual grand rounds, content sharing), 2) establishing connections and opportunities (e.g. visiting professors, patient meetings), 3) improving and utilizing a directory of clinicians and centers (e.g. criteria for identifying experts), 4) standardizing content and forms (e.g. standard intake forms, App), and 5) providing support for building coding expertise (e.g. cross-state line reimbursement of consultation). By implementing these strategies, we can continue to improve education, navigation, and consultation for those treated and affected by HPP. In 2023, we will continue working with our steering committee and working group to refine and create specific deliverables for launching our HPP Network, including an HPP patient app. 2023 will be a big year for Soft Bones and the HPP community.

## We Can Do Hard Things: Talking to Family Members About Genetic Diseases

Since HPP is a genetic disease, it is common for patients who are diagnosed to also have family members that can be impacted. Soft Bones launched a new resource on our website that provides tips on how to talk to relatives about genetic diseases, including HPP. Many patients have shared stories about how this can be a difficult and uncomfortable conversation. We worked with a therapist on the guide to help prepare for possible family dynamics and scenarios to prepare for this discussion. The guide also provides resources and support for families. Find the “HPP Talking to Your Family Guide” [HERE](#).

*Please note however, while the guide explores different scenarios, it is not meant to be a script, as conversations and communication styles can vary.*

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### LEAKY GUT, INFLAMMATION & HYPOPHOSPHATASIA

Hypophosphatasia has been known to cause increased inflammation, which is a known cause of pain. Generally speaking, anything that lowers systemic inflammation could help with pain management, and could benefit someone living with HPP.

## Follow Your Gut: The New Leaky Gut Fact Sheet

Our patients spoke and Soft Bones listened, launching a new resource on Leaky Gut, detailing important information regarding the link between gut health, inflammation, and hypophosphatasia. This fact sheet provides facts, figures, and statistics on this condition, including approaches about what can be done to help a leaky gut, top foods to avoid, and more.

To learn more, click [HERE](#).

## HPP in the News

In this year's Rare Disease Day issue of Rare Revolution Magazine, an interview with Soft Bones President and Founder, Deborah Fowler was featured. The issue focused on innovation and research and celebrates some of the amazing work taking place in the RARE community. Read the article [here](#).

Hypophosphatasia patient, Melody, discusses her HPP journey in this feature article in The Daily Journal, a local publication in the Fergus Falls area of Minnesota. Read her story [here](#).

Soft Bones was featured in a local radio interview out of Windsor, Ontario, and Detroit to talk about its mission and HPP. Listen to the interview [here](#). Keep an eye out on our social channels for a link to the interview, when available.





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## Stocking Up with Soft Bones!

### *Soft Bones Now Accepting Stock Donations*

Soft Bones is now able to accept stock donations! Donating appreciated stock is one of the simplest ways to contribute more to causes that are important to you. It's also one of the smartest ways to give to a charity. When stock donations have appreciated for over a year, contributions can be up to 20% more than if you sold the stock and made a cash donation as it helps prevent being taxed a capital gains tax. Stock donations are also an excellent way to rebalance your stock portfolio. Plus, any increase in the value of a stock can also mean significant gains for you if you sell. Donating stock to Soft Bones can be a win-win!

Please contact us at [info@softbones.org](mailto:info@softbones.org) for more info.



## Patient of the Month

Early in 2022, we launched a new social media campaign that features a new Patient of the Month. In this monthly social media post, we highlight a personal HPP story to raise awareness about hypophosphatasia and its various manifestations, shed light on the challenges patients face and provide hope to others. Patients who are featured have the opportunity to share their own HPP journey and prove to be a great way to educate friends, family members and their own communities. To read past stories, visit our website [HERE](#).

To nominate a Patient of the Month, or to share your story, email [info@softbones.org](mailto:info@softbones.org).



Nellie S. Patient of the Month  
August 2022

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# Hand in Hand: Connecting with our Youth and Young Adults

## Hippo Squad

The Hippo Squad is a program that provides HPP education for kids by providing them fun, educational activity boxes. Four times a year, children ages 3-12 rush to their mailboxes to discover what new surprises the Hippo Squad has in store for them in their quarterly box. As of 2022, more than 90 children are subscribed, and that number is continually growing. Hippo Squad boxes include a parent guide to provide instruction, oversight and background into how the activity connects to HPP. The Winter 2022 mailing was one of our most popular, featuring a baking theme with some festive holiday cookie recipes. Included in the mailing was a snowflake cookie cutter and Squad members learned that snowflakes are like HPPers in that no two are exactly the same.

To join the Hippo Squad, email [info@softbones.org](mailto:info@softbones.org)



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***Now that I'm older, I get to tell my story myself. Soft Bones Teen Advisory Council (TAC) has helped me connect with others around my age who understand what my life with HPP is like. I'm also connecting with people who know about HPP because of their family members.***

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***Davis S.***

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## Teen Advisory Council (Ages 13-21)

Soft Bones' Teen Advisory Council (TAC) is made up of more than a dozen teens and young adults, ages 13 to 21, who have been diagnosed with hypophosphatasia or are friends, siblings or children of patients. The TAC aims to provide a platform for teenagers to educate about HPP, become more involved in advocacy and fundraising, and manage content for the younger HPP community. In 2022, we welcomed several new members and that number continues to grow!



## The Strong Impact of the Teen Advisory Council

Quinn H. is a 14-year-old boy currently living with HPP. After a relatively normal first couple years of life, he started showing some gross motor delays. One of which was that he was unable to walk until he was two years old. Quinn's family didn't receive answers for his condition until a dentist suggested that Quinn might have a metabolic bone condition after he lost three baby teeth. Shortly after, he was diagnosed with HPP.



Despite being diagnosed with a rare metabolic disorder, Quinn continued to live his life to his best ability. He did not want HPP to define him or slow him down, however, he did deal with physical struggles. Quinn was finally able to find some relief when he began Strensiq, an enzyme replacement therapy. This medication has made a world of difference for Quinn. He began to feel stronger and just better overall. He has recently begun playing wheelchair basketball and softball. Quinn says it "has been a huge success" for him.



Possibly even more inspiring is Quinn's attitude and the way he approaches life. Being part of the Soft Bones Teen Advisory Council has been a way for Quinn to connect with others his age with HPP and others whose family members have HPP. Along with Soft Bones' support, he maintains that what has gotten him to this point is his positive attitude and determination to not let HPP define him. He wholeheartedly believes that no matter what life throws at you, if you get knocked down you just need to get back up and keep moving forward.





## Tooth Fairy Program

Losing a tooth can be an exciting milestone in a child's life, but for HPP families, it can be a somewhat frightening event. Every tooth lost can be a tough reminder of the challenges of living with HPP, however, for a child— a lost tooth means one thing – **the Tooth Fairy is coming!** Soft Bones has created a special Tooth Fairy Pillow featuring our HPP mascot, the hippo, that children can use when they have lost a tooth, turning this potentially upsetting experience into a happy event for everyone. We also use this opportunity to provide HPP education focused on the dental implications of the disease. Make your child feel extra special every time they lose a tooth and sprinkle a little fairy dust magic to brighten up the memory.

## Journaling My Hypophosphatasia

Soft Bones is pleased to offer a publication for the youngest members of our HPP community. If you are the parent or caregiver of a child with HPP, our children's book, *Journaling My Hypophosphatasia*, educates children about HPP and gives them a place to get in touch with their emotions as well. HPP & ME: Our Online Community This is a unique online community forum that connects patients, families and caregivers. Find answers to your questions, share stories and meet others, all in a secured environment. [Click here to register](#)



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## **We are #HPPStrong: Stories of Resilience in our Community**

Ryan is a high school junior with HPP who has a passion for all things lacrosse. He wasn't able to get the chance to play lacrosse when he was younger due to the severity of his HPP, so now he plays every chance he can get. His quality of life has improved leaps and bounds since starting Strensiq at age 12. As he got stronger, he practiced hard to try and catch up to the other kids that had been playing for years...and he ultimately made the team as a goalie! Now he dreams of playing college lacrosse. He and his mom Christine would like their extended HPP family to know, "stay strong, take one day at a time, and never give up hope that we will have a cure someday."



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## **HPP Education Never Stops: Supporting Our Healthcare Providers and Researchers**

Soft Bones' HPP Connect is the first online community dedicated to peer collaboration and knowledge-sharing in hypophosphatasia. HPP Connect is an online collaborative community for ongoing, multi-specialty, peer-to-peer exchange, dialogue and education focused entirely on HPP. It connects clinicians with experience evaluating and treating HPP to one another, fostering knowledge-sharing and collaboration that benefits patient care.



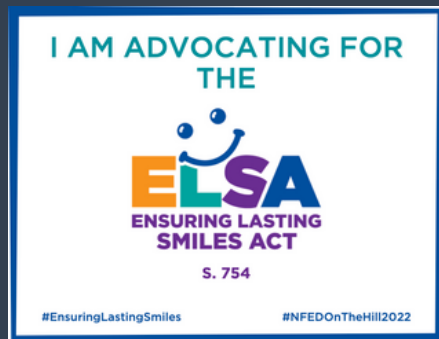
Since HPP is a rare disease that varies widely in its clinical presentation, some clinicians may have had exposure or treated only a handful of patients. HPP Connect enables access to world-leading HPP experts, allowing the medical community to exchange information and learn from one another. Ultimately, the aim is to speed up the identification and appropriate treatment of patients who have HPP. The platform allows physicians to efficiently connect, collaborate and share information in a secure, private, HIPAA-compliant online location. HPP Connect welcomes physicians involved in the direct care of HPP patients, along with residents and fellows of an accredited academic institution.



## Now Listed on Medscape

Soft Bones has been listed as a collaborator on the Medscape website. Check it out [here](#).

What is Medscape? Medscape is an online resource for physicians and healthcare professionals everywhere. They focus on the latest medical news and updates, perspectives from experts, crucial point-of-care drug and disease information, and pertinent professional education and CME courses. Overall, their goal is to advance patient care by providing healthcare professionals and physicians with access to comprehensive clinical information and resources.



## It's Time to Close The Gap: Bolstering HPP Access Support ELSA

Soft Bones is proud to have been involved in advocating for the Ensuring Lasting Smiles Act (ELSA). Under ELSA, all private insurance groups and individual health care plans would be required to cover medically necessary services resulting from congenital abnormalities. This includes services and procedures for any missing or abnormal body parts necessary to achieve normal body function, including teeth. This is an important advancement as many families without this coverage make significant financial sacrifices just to afford necessary dental care.

Soft Bones has been a proud sponsor of ELSA Advocacy Days partnering with the National Foundation for Ectodermal Dysplasias (NFED). Learn more or get involved [here](#).



Soft Bones is an active member of The Haystack Project, an initiative to amplify the voices of rare and ultra-rare disease patient advocacy organizations to highlight and address systemic obstacles to patient access. The Haystack Project's core mission is to evolve healthcare payment and delivery systems to make innovative quality treatments accessible to the patients they were meant to reach. This partnership offers our organization the opportunity to listen carefully to the specific concerns of the rare community and take action on specific actions that impact the HPP community.

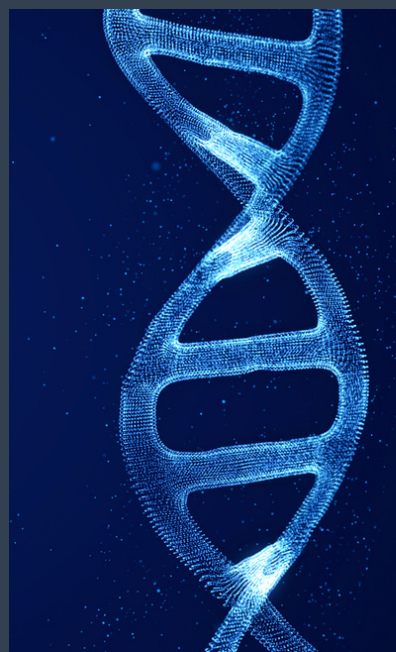
Soft Bones supported the Haystack Project's goal to pass the HEART Act (Helping Experts Accelerate Rare Treatments), which allows rare disease experts, patients, and clinicians the ability to play a role in the FDA's review process. The bill was passed by both the House and the Senate in December 2022.

## Insurance Claim Support

Navigating health insurance is extremely complex. We continue to work hard for the Soft Bones community supporting patients' medical insurance appeals. We have a deep understanding of the disease, treatments and patient needs, which can help insurance companies better understand how patients will benefit from appropriate treatment that may not traditionally be covered without clarity. Through this process, we have worked on appeals at the state level, which triggered a new possibility for new prior authorization criteria to be put in place. These small strides make big gains for those with HPP and other rare diseases.

## Institute for Gene Therapies

Soft Bones is a member of the Institute for Gene Therapies (IGT) Patient Advocacy Advisory Council (PAAC). IGT's mission is to advocate for a modernized regulatory and reimbursement framework that encourages the development of transformative gene therapies and promotes patient access. The PAAC is composed of patient advocates who represent a number of diseases and contribute important perspectives that help ensure patients' interests are at the center of IGT's work.



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## Beyond our Border: Growing Global Access

Soft Bones' efforts go far beyond our US border. We are frequently contacted by patients and physicians from outside the US who are in need of resources and we are always there to help!

### Soft Bones Has a Commitment to Sustainability

As such, we now offer welcome packets and resources in digital formats. Each year this continues to grow and we are thrilled to have a library of webinar recordings that people can access at any time. Additionally, our ECHO recordings can be accessed on HPP Connect. This further demonstrates our commitment to ensuring our resources are available across the globe.

### Diversity, Equity and Inclusion

Diversity, equity, and inclusion (DEI) is a term used to describe policies and programs that promote the representation and participation of different groups of individuals, including people of different ages, races and ethnicities, abilities and disabilities, genders, religions, cultures, and sexual orientations. Hypophosphatasia does not discriminate, and its effects can be felt by people of all genders, ages, races, ethnicities, socioeconomic statuses, religions, and sexual orientations. As a rare patient population, we have experienced first-hand the impact of what it feels like to be excluded, discounted, and ignored.

As such, the mission of Soft Bones is to provide a place where HPP patients feel that they are not alone, which means our staff is committed to creating an environment that is free from discrimination, where everyone is welcome and people come to connect, learn and ultimately thrive. Our DEI work takes on many forms, from incorporating diverse viewpoints from patients with various levels of severity of HPP to connecting with researchers across the globe, from different backgrounds and cultures to ensuring our materials are available in many different languages and in hard copies as well as online. To date, Soft Bones has translated materials into Arabic, Polish, Portuguese, and Spanish, with many more international languages on the horizon to further the accessibility of resources for those who need them most.

Every person deserves to be listened to, respected, and have access to high-quality healthcare. We are committed to helping all families improve their understanding of hypophosphatasia and build their health literacy in order to become more informed consumers of the healthcare system and become empowered to access the critical care their loved one needs and deserves. HPP isn't confined to the borders of a country - so access to quality care, support, and educational materials shouldn't be.



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## 10th Maher Family Grant awarded to Nan Hatch, DMD, PhD

As an organization committed to bringing forward advancements and education to those affected by HPP, it was an honor to award the 2022 Maher Family Research Grant grant to Nan Hatch, DMD, PhD. As an associate professor of dentistry at the University of Michigan School of Dentistry, this \$25,000 grant will support Dr. Hatch's studies to establish essential roles for tissue-nonspecific alkaline phosphatase (TNAP) in muscles and determine how TNAP modulates muscle function. Dr. Hatch's previous findings led her to question if cell cycle and/or energetic abnormalities might also be relevant to the muscle weakness and fatigue found in some individuals with HPP.



Nan Hatch, DMD, PhD

Soft Bones funds research to further the understanding of HPP, focusing on finding a cure. To date, Soft Bones has awarded over \$475,000 in research grants since 2014. The organization supports HPP investigators through annual grants such as the Maher Family Annual Hypophosphatasia Research Grant.

More information about hypophosphatasia, as well as existing and ongoing research can be found [HERE](#).

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## Gene Therapy Update

Soft Bones has made it a priority to focus on new therapeutic approaches to treating HPP, including gene therapy. Research from Sanford Burnham Prebys highlights [a new study that shows promising results for those with HPP](#). Published in the Journal of Bone and Mineral Research, this study was performed in a mouse model of the disease, further supporting the advancement of gene therapy toward human clinical trials. This preclinical lab highlights Soft Bones' Scientific Advisory Board member, Dr. José Luis Millán, along with other scientists at Sanford Burnham Prebys, established that AAV8-TNAP-D10--a gene therapy that replaces a key enzyme found in bone--may be a safe and effective single-dose treatment for hypophosphatasia (HPP). Dr. Millán was the recipient of our Soft Bones grant, and his lab was funded in part by this grant.

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## The Growth of our our Board

In 2022, we added three more esteemed members to our Scientific Advisory Board and also one to the General Board of Directors. Welcome to HPP experts Drs. Kathryn McCrystal Dahir, Vanderbilt University Medical Center, Gary S. Gottesman, Washington University School of Medicine, and Peter Tebben, Mayo Clinic to our Scientific Advisory Board and Rob Moskow to our Board of Directors.

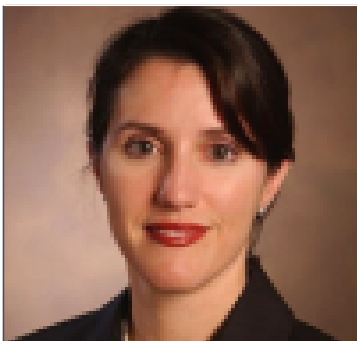
Our Scientific Advisory Board will help guide the way for our initiatives and provide expertise in their specialty areas of practice and research. To learn more about each of the three professionals, visit the Scientific Advisory Board section in the ['About Us'](#) section of the Soft Bones' website.

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Soft Bones powers the HPP community by raising awareness of this rare, metabolic disease, providing hope by advocating for and funding research, and connecting patients with each other and with the medical community to advance the understanding of HPP.

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**Kathryn McCrystal Dahir**  
MD, Vanderbilt University Medical Center



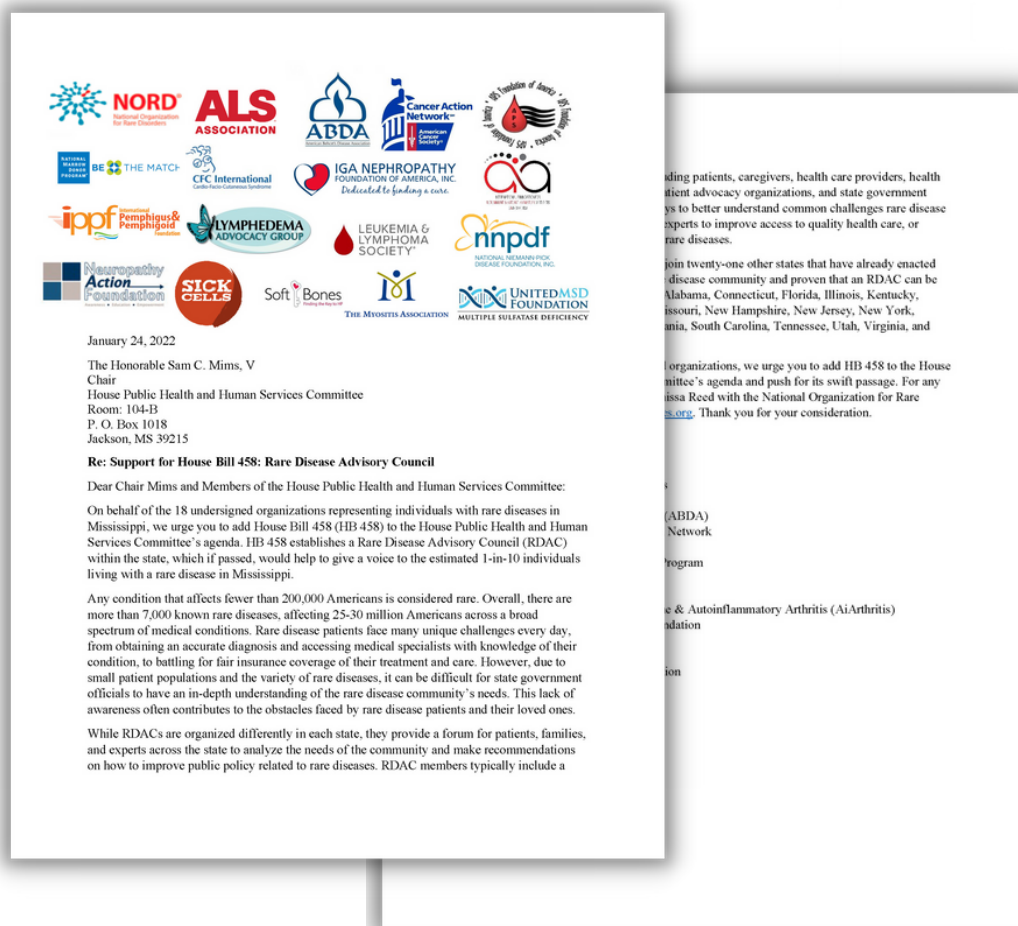
**Gary S. Gottesman**  
MD, Washington University School of Medicine



**Peter Tebben**  
MD, Mayo Clinic

## Joining Forces in Support of Mississippi Rare Disease Advocacy Council

Thanks to hypophosphatasia patient and advocate Aaron Blocker, Soft Bones became aware of an opportunity to join the Mississippi Rare Disease Advocacy Council. On Friday, January 21, 2022, Soft Bones, along with 17 other organizations representing individuals with rare diseases in Mississippi, signed a letter of support urging the Honorable Sam C. Mims, Chair, and Members of the House Public Health and Human Services Committee to add House Bill 458 (HB 458) to the House Public Health and Human Services Committee's agenda. HB 458 establishes a Rare Disease Advisory Council (RDAC) within the state, which if passed, would help to give a voice to the estimated 1-in-10 individuals living with a rare disease in Mississippi. To learn more, read the full document [here](#).







**\$188,712.20.**

## **Fore The Cause: The 2022 Soft Bones Golf Classic**

The 14th Annual Soft Bones Golf Classic was held on September 19th at Somerset Hills Country Club in Bernardsville, NJ. This is our largest fundraiser, and this year's golf classic was another successful event. We had 113 golfers join us for another beautiful fall day. Sponsors, donors, patients and participants had a great time playing the course while helping raise critical funds for our organization. We are grateful for the loyal golfers who turn out year after year to support our mission. A special thank you to our Tournament Sponsor, Atlantic Health System.



**Tournament Sponsor:** Atlantic Health System

**Birdie Sponsor:** The Fowler Family Charitable Foundation

**Eagle Sponsors:** Donnelly Construction & Gates and Mary Ellen Hawn

**Putting Green Sponsor:** Berkshire Hathaway AZ - Harvey & Linda Salkow & Andrew & Ann Stock

**Beverage Station Sponsors:** William F. Jones, DMD

**Additional Tee Sponsors:** Acorn Financial Services, Kenneth Banasiak, DMD, Greg and Amanda Boring, Christ the King - New Vernon, NJ, Connect One Bank, The Fowler Family, John Kenney, Henry Kim, M&M Perrotti's, Dan & Dani McNeil, McRae Capital Management, The Mulcahy Family, The Murphy Family, Natirar, Stephanie and Swan Oey, The Richardson Family, and Wendy Supron.

**19th Hole Sponsors:** Miller Bugliari, Francis Kenneally and Joseph Micale, DMD



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## In Memoriam: Bob Mulcahy, Chairman of the First Soft Bones Golf Classic



It is with heavy hearts that we share the news of the passing of Bob Mulcahy. The 2022 Soft Bones Golf Classic was our first without Bob as Chairman of the event. As many know, Bob stepped up more than 14 years ago when Cannon Sittig was first diagnosed, and Deborah Fowler talked about the need of starting a patient advocacy group for families living with hypophosphatasia.



He was committed to the cause, and through his incredible network, helped raise more than 1-million dollars for Soft Bones, making much of what has been accomplished possible. The Soft Bones Golf Classic remains the organizations #1 fundraiser and will continue in the generous spirit that Bob brought to the event. Click [here](#) to read more about Bob and the legacy he leaves behind in New Jersey. He will forever be missed.

# Fundraising Highlights



**24** people created Facebook fundraisers raising **\$11,457.37**

plus many more who pushed the donate button etc.  
Thanks to those who used Facebook to create online fundraisers in honor of special occasions like birthdays, anniversaries and special dates (Rare Disease Day, World HPP Day etc.)

**\$2,214.08** dollars raised via Amazon Smile



## Online Shopping Totals:

- Little Caesar's- **\$672.00**
- Pampered Chef- **\$302.95**
- Krispy Kreme- **\$588.35**
- Wreath Fundraiser- **\$85.17**

**Pretty Pink Rooster**  
Shopping Fundraiser- **\$236.97**

**Enriched Aesthetics**  
Botox and Bubbly Fundraiser- **\$400.00**

## Additional Fundraising Highlights

Rutgers Men's and Women's Lacrosse Fundraiser Recap - The Rutgers Men's and Women's Lacrosse teams partnered together to raise money for Soft Bones as part of their annual charity event. The combined value raised by both teams was **\$6,691.50!**

SpreadShop is an online logo shop linked to our website where a percentage of proceeds benefit Soft Bones. Specialty items are added during important campaigns and events such as, World HPP Day, where a portion of proceeds from item sales are donated back to our organization. Additionally, the Soft Bones Teen Advisory Council (TAC) created their own t-shirt design to help generate sales and raise funds. Each year, a shirt design is created for our largest patient meeting of the year, the National Patient Meeting which also raises funds through a portion of sales. In 2022, the SpreadShop campaign sold 313 items and earned **\$1,486.49** in sales!



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## Cannonball Run Fundraiser

This year, Soft Bones hosted its first-ever 5k: The Cannonball Run, Roll, or Stroll! During the month of October, participants were urged to complete their 5k by running, rolling or strolling, any time, anywhere! Some members of our community were even able to complete it together. With 67 registrants, the HPP community came together to bring global awareness to hypophosphatasia. In total, we raised **\$1,675.00** and an additional **\$1,006.50** in donations. Soft Bones would like to extend a huge thank you to all of the participants.

## Cannonball 5k



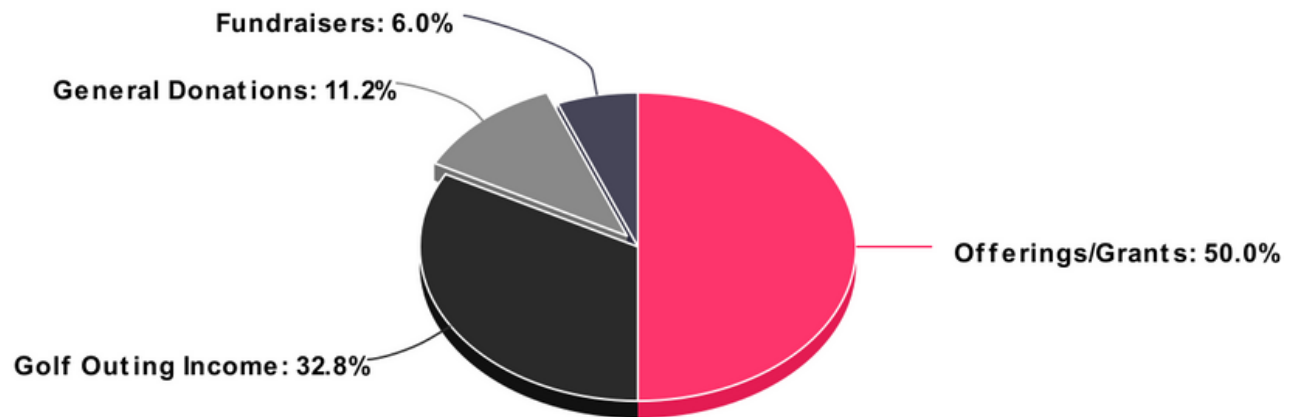
**Run, Roll, or Stroll**



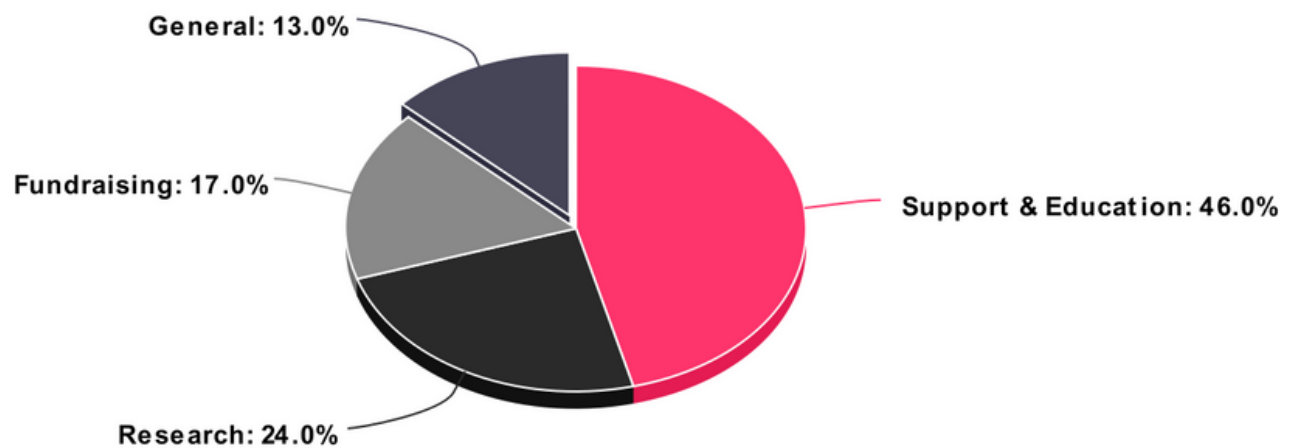
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# Key Financial Information

## Donation Campaigns



## Expenses





# How You Can Help



[Volunteer](#) | [Donate](#)





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# List of Donors

## Cannonball Run

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Rachael Alford  
Pat Canada  
Dominic Carrano  
Maryanne Caruso  
Jane Castello  
Kristi Culver  
Brenda Degnan  
Laura Ekas  
Linda Eramian  
Catherine Fenlon  
Jennifer Flanagan  
Deborah Fowler  
Jason Fowler  
Caroline Fox  
Kim Fox  
Ryan Fox  
Carolina G.  
Bill Goodbar  
Chloe Goodbar  
Denise Goodbar  
Jim Grady  
Michael Greco  
Judith Harris  
Amanda Hayward  
Claire Hernon  
Megan Hessel  
Ann Houston  
Daina Janowski  
Kelly Jones  
Noel Lammey  
James Latimer  
Laiken Latimer  
Preston Latimer  
Pamela Legett  
Eric Levesque  
Caden Lucarelli  
Kinsley Lucarelli  
Brad MacDougall  
Hillary MacDougall  
Daniela Martinez  
Seametso Maseng  
Debbie McGarity  
Charlie McGarity  
Todd McGarity  
Kirsten Mitchell  
Mary Mulcahy

Lila Namisnak  
Maya Namisnak  
Pam Neumann  
Deana Ollis  
Keith Ollis  
Tricia Oppelt  
Allison Pishko  
Traci Raynor  
Cindy Reasor  
Christine Salomon  
Nellie Sanders  
Cannon Sittig  
Clark Sittig  
Colby Sittig  
Grace Sittig  
Travis Sittig  
Connie Tavanis  
Samantha Tracy  
Ting Yi  
Briley Zibilski  
Danielle Zibilski  
Grayson Zibilski  
Travis Zibilski

## Golf Classic

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Charles & Karen Ann Baracco  
John & Mary Burke  
Dan Burkhart  
Peter Cannenbaum  
Cara Dahms  
Margaret & Richard Depaul  
Bob & Amelia Doherty  
Heidi Floyd  
Joseph Guadio DDS  
John & Judith Harris  
Deidre Hart  
Thomas Hughes  
James Kavanaugh DDS  
Jack Ketterson  
Samrat Khichi  
David Kulick  
Robert Leeks  
Brian Maher  
Kevin Mulcahy  
Mary Mulcahy  
Francis Romano III & Megan Mulcahy

Thomas Sheridan  
Dr. Stephen Verdasco &  
Patricia Durner  
Maragert Warner  
Amy Weinberger  
Forest & Margaret Willaims  
Cathleen Witczak

## Online Fundraisers\*

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Stephen Augustyn  
Margo Bottilla  
Nicol Busby  
Melinda Cherry-Proudfoot  
Shawn Cole O'Ryan  
Kristin Cook Pierce  
Deborah Fowler  
Olivia Hasko  
Kathie Haupt- Britt  
Brianna Hines  
Deziree Holaday  
Jennifer Jamison Taylor  
Lorrie Leaf  
Jessica Leaf  
Amanda Loggins Ramey  
LaVonne McCombie  
Kirsten Mitchell  
Vicki Orr  
Lauren Rasmussen  
Jill Riggan Acton  
Kenneth Rossen  
Diane Theroux  
Myra Kay Waltz  
Danielle Zahm-Zibilski

## Pampered Chef

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Susan Brown  
Michelle Hutson  
Alison Purdon  
Cindy Reasor  
LuAnne Rutter  
Sharon Talkington  
Sheila Zimmermann

## Norwex

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Ann Haak

## Little Caesar's Fundraiser

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Amy Weinberger  
Cindy Reasor  
Blynda Kellner

## End of Year Giving (Giving Tuesday & Annual Appeal)

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Virginie Bitterlin  
Marie Boo  
John Braniff  
Charles & Noreen Bruns  
John Callandrello  
Ashley Carbone  
Hillma Denune  
Bob & Amelia Doherty  
Christine Douglas  
Marilee Falde  
Kathleen Fleschner  
Heidi Floyd  
Judith Janansky  
Shirley Kengott  
Laiken Latimer  
Sharon Martin  
George (Jr.) & Jeannie Meeks  
Charles & Betsy Miller  
Alek Miller  
Dobrawa Napierala  
Robert & Mary Jane Nettune  
Grover Owens  
SueAnn Peete  
Robert Perrault  
Minerva Richardson  
Julie Ried  
Patricia Romano Wolf

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## End of Year Giving (Giving Tuesday & Annual Appeal continued)

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Patricia & Joel Romano Wolf  
Farris & Sherry Sittig  
Leann Slezak  
Royer Smith  
Mick & Tonya Syslo  
Stephanie & Perry Troisi  
Margaret Vernet  
Victoria Wideman  
Lindsey Willson  
Rosemarie Yancosek  
Barbara Young  
Green Earth Financial  
Professionals LLC  
G. Stone Connections, L.L.C.

## Donors: General

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Matt Adey  
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Caren Begun Wagner  
Erika Beyer  
John Braniff  
Becky & Ken Breedon & Family  
John Callandrello  
Ashley Carbone  
Thomas Carpenter  
Elizabeth Carter  
Kevin & Suzanne Carton  
Paige Clark  
Chip Clements  
Julie Cole  
Chris Connors  
Robert Densley  
Richaerd Derrick  
Kuljit Dhinjal  
Emma Farwell  
Sandra Felberg  
Malcolm Forbes  
Kathleen Fowler  
Brendan Fowler

Richard Fowler  
Kim Fox  
Anna Frable  
Robyn Gay  
Judith M. Greene  
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Richard Hanlon  
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Chrystal Moynahan  
Patrick Mucci  
Kevin Mulcahy  
Aaron Perry  
Margaret Ranis  
Kathy Reagor  
Patricia & Joel Romano Wolf  
Laura Rubenstein  
James, Tamyra & Wes Ruzicka  
Michael & Nellie Sanders  
Sargento Foods Inc  
Elaine Stafford  
Mary Tedrick  
Deborah Wenkert

## Donors: In Honor/ In Memory

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Jaime Ballard  
Marie Boo  
Britt Family  
Michael & Jennifer Caputo  
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Loree Carter  
Michael & Sharon Chittick  
James & Laureen Dillon  
Mary Lou Dobrydnia Pod  
Mary Donohoe  
Kathy Edwards  
Bonnie Enfinger  
Garry & Joanna Finch  
Linda Glavin

John & Carole Landsverk  
Daniela Martinez  
Julie Ried  
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Allen & Sherri Rosen  
Patricia & Stephen Shaw  
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