

# THE BARE BONES

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

## Soft Bones Awards Grant to The Ohio State University College of Dentistry Researcher



**Fatma Mohammed, Ph.D.**

Our Foundation was pleased to award the 9th Annual Maher Family Grant to Fatma Mohammed, Ph.D., a postdoctoral scholar in the Dr. Brian Foster Laboratory at The Ohio State University College of Dentistry. Dr. Mohammed was awarded a \$25,000 grant to support her research on the interactions between dental, skeletal, and nervous systems on bone and teeth defects seen in HPP. Through this research, Dr. Mohammed hopes to gain new insights on how HPP impacts bone and teeth and how this relates to disease manifestations outside the musculoskeletal system.

Soft Bones awards annual grants such as the Annual Maher Family Grant, for research that is directly related to hypophosphatasia.

Read the full announcement [here](#).

### IN THIS ISSUE:

- Message From Our Founder
- Rare Disease Day 2022
- Project ECHO Update
- HPP Connect Collaboration



## New Year Message From Our Founder

**Dear HPP Community,**

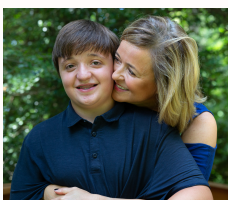
I don't think any of us expected 2021 to pack such a punch. COVID-19 variants continue to impact our community and the world at large as we enter the new year. Living with a chronic disease during a pandemic brings its own set of challenges. However, our HPP community knows how to be resilient and carry on in the face of uncertainty. With a new year comes renewed hope for brighter days. Since we founded Soft Bones more than a decade ago, we also have learned the importance of collaboration, connection, and knowledge sharing for tackling difficult obstacles and identifying solutions that enable all of us to live our best lives.

Despite the challenges of the pandemic, we continued to offer our HPP families informative educational programs in virtual formats in 2021, including our first-ever virtual patient meeting and regional patient meetings. Our 2021 World HPP Day theme, "Let's All Be Heard", took on new importance as we raised our collective voices for HPP awareness, proving to ourselves that we are truly stronger together. Through our TeleECHO program, we educated healthcare professionals about HPP by offering presentations and discussions on different HPP-related topics, enabling them to amplify the knowledge for providers to deliver best-in-class care in their own communities. We exhibited virtually at the American Society for Bone and Mineral Research (ASBMR) Annual Meeting, awarded our 9th annual research grant, and announced our partnership with Aruvant Sciences making our first gene therapy research for HPP. And, our fundraising programs raised vital funds for education, advocacy, and research to benefit our organization and community.

I am grateful to everyone who contributed their time and energy to support our organization's goals, especially during these difficult times. Thank you to those who supported our foundation with a donation. And a special thank you to our Region Leads for keeping an ear to the ground at the community level to support the needs of patients and ensure their voices are being heard.

As we look ahead, we are so excited about some wonderful new initiatives we're advancing for both patients and providers that are described in this newsletter. We continue to expand our offerings to meet the needs of our community, address knowledge gaps and support HPP research and enhanced patient care. We are also looking to once again gather in person in Orlando for our patient meeting in July. Clearly, your health and safety are our top concern and we will keep a close watch on the CDC's guidance for travel and gatherings. I am hopeful our meeting will continue as planned, and I can't wait to see all of you in person.

My best wishes to you and your loved ones in 2022.



**Sincerely,**

**Deborah Fowler**  
**President and Founder**





## Recap: World Hypophosphatasia Day 2021

This past October, we came together as an HPP community to raise global awareness of hypophosphatasia, culminating on **World Hypophosphatasia Day (October 30)**. This year's theme was "**Let's All Be Heard**," which provided an opportunity for patients, caregivers, and supporters to share their stories while bringing awareness to the unseen emotional side of daily life with HPP. Thank you to everyone who participated by telling their stories and getting involved both online and in their local communities.



Thanks to our fundraising partner, HundredX Causes, and our HPP community, Soft Bones raised \$5,574 during the World HPP Day online feedback fundraising campaign!

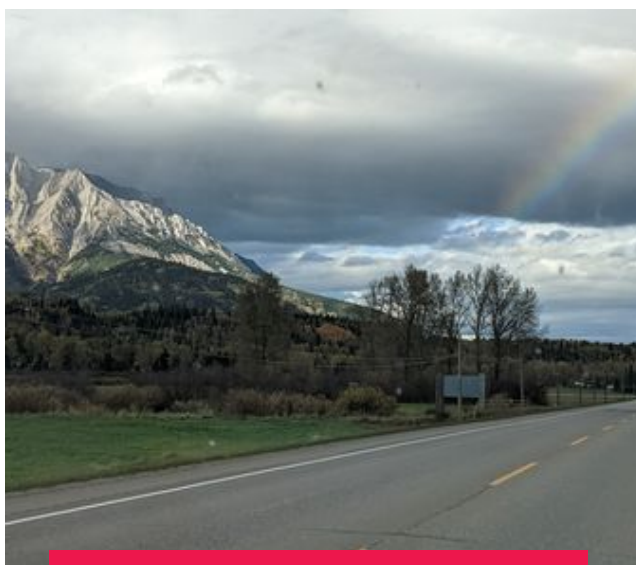
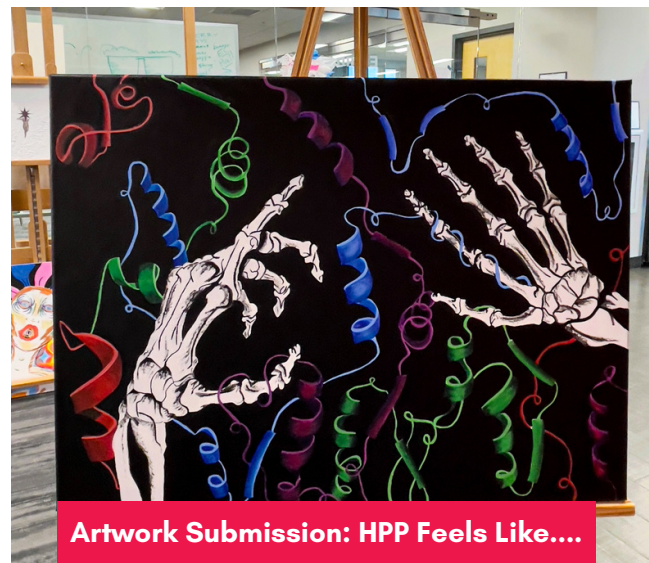


Photo Submission: HPP Feels Like....



HPP Community Halloween Virtual Chat

To learn about our impact, read the full recap of activities and initiatives [here](#).





## Countdown to Rare Disease Day 2022: Share Your Colours

February 28th is Rare Disease Day- and we're joining the many organizations around the world to bring awareness to the rare diseases that impact over 300 million people globally.

**Here are some things you can do from home and in your community to show support for Rare Disease Day and our HPP family:**



- **Wear it loud and proud!** Let people know you're a rare disease warrior with our new t-shirt design, available now in the [online store](#). Order yours today so it will arrive just in time to wear on RDD.
- To bring awareness to the wide range of symptoms that affect patients living with HPP, **download and complete [this coloring activity sheet](#)** and share it with your friends and family on social media. You'll be entered to win a Soft Bones hippo- see article below!
- **Tag us in your posts** (@SoftBones- Facebook or @SoftBonesHPP- Instagram) and use the hashtags #SoftBonesHPP and #RareDiseaseDay and you'll be entered to win some Soft Bones swag! Winners will be announced on March 1. See further contest rules below.
- Join us for our next webinar on Rare Disease Day (February 28th): **"What is Gene Therapy? - Its Promises and Challenges for HPP."** This webinar will cover: what it is, what it isn't and what it could mean for HPP. **[Register today!](#)**

To learn more about Rare Disease Day and how you can get involved, visit [www.rarediseaseday.org](http://www.rarediseaseday.org).

## Tag Us This February: Win Soft Bones Swag!

**#ShareYourColours this Rare Disease Day, February 28, and you could win our sought-after Soft Bones hippo!**

### **How It Works:**

- Download and complete the **[Soft Bones coloring activity sheet](#)**- children and adults welcome to participate!
- Snap a pic of your design and share it on social media
- Tag us in your post (@SoftBones- Facebook or @SoftBonesHPP- Instagram)
- Use the hashtags #SoftBonesHPP and #RareDiseaseDay

**Winners will be announced on March 1, 2022.**



## Coming Soon: New HPP Research Announcement

**HPP PATIENTS AND CAREGIVERS:** There's a new opportunity for the HPP community to participate in new dental research through The Ohio State University. The announcement will be sent out to patients by CoRDS through the [HPP Patient Registry](#). To join the CoRDS HPP Registry, click [here](#).

**CoRDS**  
Rare Disease Registry



## Update: HPP TeleECHO For Clinicians and Researchers



Zoom Meeting  
Recording

**HPP Is Caused by Mutations in the Gene Encoding Tissue Non-Specific Alkaline Phosphatase (TNSALP)<sup>1</sup>**

**Mechanism of Disease<sup>2</sup>**

```

graph TD
    A[Autosomal recessive or dominant Mutation(s) in ALP Gene Encoding TNSALP] --> B[Low ALP Activity]
    B --> C[Substrate Accumulation]
    C --> D["PPi*, PLP, PEA"]
    D --> E[Skeletal & extra-skeletal manifestations of HPP]
  
```

ALP: alkaline phosphatase  
PPi: inorganic pyrophosphate  
PLP: pyridoxal-5'-phosphate, biologically active metabolite of vitamin B6  
PEA: phosphoethanolamine

<sup>1</sup> Whyte MP, 2000; <sup>2</sup> Rockman-Greenberg C, 2003; <sup>3</sup> Mornet E, et al, 2016.

**Dr. Dahir presents her session on Adult HPP via Zoom**

We are thrilled to announce that after completing the first six sessions of our HPP TeleECHO series, we will be continuing the program for six more months. The series was well-received, with an average of 98 registrants per session. The TeleECHO faculty, including Drs. Michael Whyte, Shriner's Hospital For Children- St. Louis, Matthew Drake, Mayo Clinic, and Kathryn McCrystal Dahir, Vanderbilt University Medical Center, have identified the topics and speakers for the remaining sessions, which will begin in February 2022.

(Continued from pg. 5) The Soft Bones HPP TeleECHO offers the opportunity for HPP experts to share their knowledge with other physicians and researchers who would like to expand their knowledge base as they treat current or future HPP patients. If you think anyone on your healthcare team could benefit from attending these sessions, feel free to share a program flyer with them at your next appointment, or email [ECHO@softbones.org](mailto:ECHO@softbones.org). Thank you to those of you who recommended your dental care professional to receive a flyer for the December session. To view the program flyer and a list of topics covered, click [here](#).

## New Members Offer Expertise To The SAB

Soft Bones is thrilled to announce the addition of three esteemed members to our Scientific Advisory Board. 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!**

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 on the **'About Us' tab** of the Soft Bones' website.

### WELCOME TO OUR NEW SCIENTIFIC ADVISORY BOARD MEMBERS



**PETER TEBBEN, MD**

Mayo Clinic



**KATHRYN  
MCCRystal  
DAHIR, MD**

Vanderbilt University  
Medical Center



**GARY S.  
GOTTESMAN, MD**

Washington  
University School of  
Medicine



## Soft Bones Launches Collaboration Platform for Health Professionals

In December, Soft Bones launched the first and only 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.

Because 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 experts in HPP, 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, HIPPA-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 and researchers interested in HPP. For physicians and researchers who would like to learn more, email [HPPConnect@softbones.org](mailto:HPPConnect@softbones.org).

**Patients and caregivers-  
please share this flyer with your  
healthcare providers.**



## Reminder: Changes to Facebook Groups Effective February 1st

Attention HPP Facebook Community members: don't forget- some of our Facebook groups are moving to our more secure, easily searchable online platform, **HPP AND ME**.

### You asked and we listened!

By February 1st, Facebook groups for each U.S. region, along with the Hypophosphatasia (HPP) Girls Cafe and Alternative Therapies to HPP groups, will be closed, and important conversations will be moved to the HPP AND ME platform, which can be found under the "community" section of the Soft Bones website. HPP AND ME is an online forum for diseased-focused conversations and exclusive first looks at new Soft Bones, Inc. content and breaking news.

With continuing concerns voiced from patients about privacy and the growing number of social channels, HPP AND ME allow patients to communicate about a variety of topics in a single platform. Consolidating the conversations will allow Soft Bones staff to focus on other priorities for the HPP community. All Soft Bones Region Groups on HPP AND ME will be password-protected for privacy. If you need the password, reach out to [Cindy@SoftBones.org](mailto:Cindy@SoftBones.org).

In addition, the Facebook private group "Patients and Caregivers" will remain open, and can still be used for Good News Friday, sharing news stories and connecting with others in the Soft Bones HPP community. HPP AND ME was created for diseased-focused conversations, a place to share experiences, discuss injection site issues, and additional complications you or your loved one might be facing.

## Soft Bones to Host First Patient-Focused Drug Development Meeting

Our foundation is planning its first-ever Patient-Focused Drug Development (PFDD) meeting to be held later this year. Using a process established by the U.S. Food and Drug Administration (FDA), PFDD meetings are designed to better understand the patient perspectives on specific diseases and currently available treatments. The FDA encourages patient organizations to hold PFDD meetings, bringing together patients, care partners, doctors, and others to examine how a condition like HPP impacts daily life.







(Continued from pg. 8) Through our PFDD meeting, we hope to characterize the HPP patient experience and identify knowledge and treatment gaps, all informed by the HPP community. The meeting will capture important insights about HPP disease progression and the disease burden for patients, families, and caregivers. It puts the patient voice front and center since people living with the condition are often the ones with the most knowledge of and experience with HPP.

The public input we generate through our PFDD will potentially be published on the FDA's website, ensuring that HPP patient experiences, perspectives, needs, and priorities are captured and documented. Soft Bones will be able to meaningfully incorporate insights into future research and policy that improve patients' lives and make a meaningful difference in their treatment and care, both now and in the future.

Stay tuned for updates and more information.

## Teen Advisory Council Update

The TAC has had an exciting couple of months! After launching a Venmo bingo board fundraiser for World HPP Day, the group raised \$307!!! They practiced active advocacy and the utilization of social media and raised far more than the anticipated goal. Together, they created a t-shirt design which is now published on the Soft Bones website- you can grab yours today!

The group is excited to show off their "official" merchandise in public.

Soft Bones will be opening enrollment for the TAC from January 30th until March 18th. Members of the HPP community, siblings, and friends ages 13 to 21 are invited to submit an application to join an amazingly passionate group of teens.

*Provided by TAC Coordinator Julia O'Leary*

**SUBMIT YOUR  
APPLICATION  
TODAY**



## Advocacy Update: Soft Bones Joins 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](#).

## Welcome New Staff Members

Soft Bones would like to welcome three new team members to our internal staff: **Emma Farwell, Marketing Specialist, Cindy Reasor, Project Coordinator, and Liz Frawley, Administrative Assistant**. As we grow, we are excited to continue supporting the HPP community. Please take a moment to welcome them to the Soft Bones team!



Emma Farwell



Cindy Reasor



Liz Frawley

## Join Us For The 2022 National Patient Meeting

This important event will bring together HPP community members from all over the United States to gain valuable knowledge while connecting with others. The meeting will include updates and education sessions presented by healthcare specialists and Soft Bones staff, as well as bonding activities for adults, children and teens. Those who have connected online but haven't met in person will also have an opportunity to do so.



Whether you were able to come to one of our past meetings, or you'd be coming for the first time, don't miss the opportunity to experience a LIVE Soft Bones National Patient Meeting! Presentations will be made available online immediately following the event for anyone who cannot attend in person, or would like to rewatch the sessions.

Learn more at [www.softbones.org](http://www.softbones.org). Registration will be open very soon!

## Upcoming Event

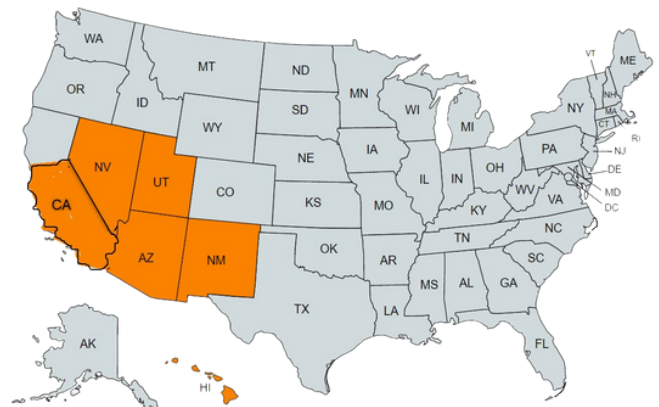
### Southwest Region Virtual Chat

Saturday, February 5th

12pm PST

Join us online via Zoom for an afternoon of conversation and friendship as we share stories and experiences with HPP. Open to all patients and caregivers in the states highlighted.

**> REGISTER NOW**







## New Resources Round-Up

The Soft Bones website offers a comprehensive [online library](#) of webinars, fact sheets and other resources that can be easily downloaded by patients and caregivers, as well as printed or shared via email with physicians and healthcare specialists. Educational materials are helpful tools to bring to doctor's visits when being treated for a rare disease like HPP. As information constantly evolves, new resources are always in development and being added, so check back often!

- Soft Bones now offers a valuable resource for expectant parents and families with babies and young children affected by HPP, the **New Parent Guide to HPP**. This comprehensive piece outlines a range of topics that are of importance to parents and caregivers during the early stages of diagnosis when information can be overwhelming and quite confusing. The document is available for download or in hard copy for caregivers/families, as well as healthcare professionals for use in their practice. We would like to give a special thanks to all of the families who shared their stories and provided feedback to the guide, and to the Committee who helped develop content, Dr. Gary Gottesman, Dr. Eric Rush, and Dr. Jill Simmons.
- Pain is an unfortunate yet common symptom of hypophosphatasia for a variety of reasons. This informative fact sheet, **Managing Pain in Hypophosphatasia**, developed in partnership with Christopher Sobey, MD, of Vanderbilt University Medical Center, examines the sources and impact of pain on individuals and offers strategies that help minimize its effects.
- **Newly released educational sessions**– see next page (13) for the complete list of recordings now available from the recent Midwest 1&2 Region Meeting, which can be found in our website's **"Resources" section**, under "Webinars."





## HPP Community: Region Updates

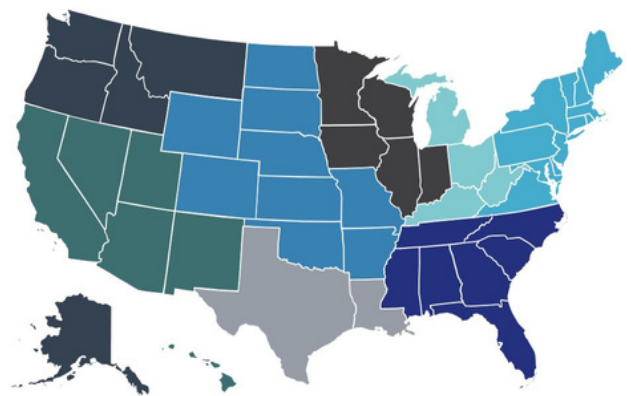
### **Patients and caregivers- have you participated in a regional activity yet?**

Gain peer support and learn about your local resources by attending a virtual event and connecting with your local region lead today!

### **A special thank you to the region leads for hosting the following meetings this quarter.**

Each meeting was led by the Region Lead (volunteer patient/caregiver) for the area, and included presentations by qualified healthcare professionals and/or guest speakers:

- **Northeast-** led by Region Lead Chris Denune, which included an excellent overview of physical therapy and movement-related implications of HPP by Donna Griffin, PT, DPT, PCS, Shriners Hospital For Children- St. Louis, and discussed current research being conducted for potential HPP Centers of Expertise through a focus group led by Consultant Gloria Stone.



- **Midwest 1&2 - Combined Event-** led by Region Leads Sharon Talkington and Haley Gast, which featured presentations on the Journey to HPP Diagnosis by Steven Ing, MD, The Ohio State University Wexner Medical Center and Pamela Brock, MS, Licensed Genetic Counselor, The Ohio State University College of Medicine; Foot Ailments Related to HPP by Said Atway, DPM, The Ohio State University Wexner Medical Center; Intro to Tooth Development and Dental Tissues by Brian Foster, Ph.D., The Ohio State University College of Dentistry; Orthodontic Treatment for the Growing Patient by Kaitrin Kramer DDS, Ph.D., Nationwide Children's Hospital; Prosthodontic Treatment in Individuals With HPP by Damian Lee, DDS, MS, FACP, The Ohio State University College of Dentistry; and an introduction to the OneSource Program by Heather Minore-Cotto, Nurse Case Manager, Alexion, AstraZeneca Rare Disease. The recorded meeting can be found within our website's ["Resources" section](#), under "Webinars."

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



## Fundraising Corner

THANK  
YOU

**to the entire Soft Bones Community for supporting our  
Year-End Giving Campaign!**

**With your help, we exceeded our goal and received an  
additional \$10,000 match from an anonymous donor!  
We can't thank you enough! Your support helps us move the  
needle towards our ultimate goal: to find a cure for HPP.**

**A heartfelt thank you to all of our community fundraisers who hosted an event or  
created a fundraising page to benefit our organization this quarter:**

### **Pampered Chef Virtual Hostess Party:**

Sheila Cane

pampered|chef.

### **Facebook Fundraisers:**

Blynda K.

Sebastian W.

Joslyn K.

Amberleigh

Sue K.

Sandy

Laiken L.

Kenneth R.

Mela N.

Mindy

Melissa B.

Becky R.

Anthony M.

Brianna

Sam

Kara G.

Anna W.

Liv T.

Sealey S.

Alexander P.

Brenda P.

Johnny K.

Amy B.

Diane T.



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