Soft Bones
Finding the Key to HPP

SPRING 2017

SOFT BONES ON CAPITOL HILL

Soft Bones was privileged to support the American Society of Bone and Mineral Research (ASBMR) in its annual "Hill Day" to raise awareness of the bone organization and to advocate for issues important to our HPP community. It truly was a great day, meeting 5 members of Congress.
It was also a learning experience as there are a couple of key issues right now that impact HPP. First, Congress is currently operating under a contining resolution (known as a CR). The CR is a stopgap to prevent a government shutdown while a final budget is being negotiated and passed. The reason this is important is because it directly impacts the funding of the National Institutes of Health (NIH) by putting research grants, and any future funding, on hold. Since many HPP scientists benefit from NIH funding and grants, we lobbied to urge congress to work together to get a budget approved by April 28 to avoid another CR.
We also met with the Congressman Leonard Lance who currently co-chairs the Rare Disease Caucus and spoke about the needs of HPP patients who are part of the ultra-rare population -- which has a different set of needs and considerations. For example, some healthcare legislation places lifetime spending caps on rare disease patients. Knowing the costs of Strensiq, this type of legislation could be very damaging to the HPP community.
We also met with Representative Frank Pallone's staff office who is active in FDA process and how to better bring patient insights into drug approvals. We talked about Sue Krug's role as a patient representative and they were really excited to see we were already engaged. We also offered to be an active resource to the government to inform them of the needs of the HPP/Rare Disease Community.
Most importantly, we did what we do best to "connect the dots" by informing lawmakers about the needs of the research community, patients, and then taking their feedback with us to better inform our communications and plans moving forward.

SOFT BONES RESEARCH UPDATE

Soft Bones is committed to promoting the development of innovative research by new and established investigators in HPP. As a result, Soft Bones has awarded a Research Grant for the past three years and will do so again in 2017. This is a seed grant of $25,000 for basic or clinical research directly related to HPP.

Thank you to our past winners for providing us with an update on their exciting research.

2014 Recipient - Steven Mumm, Ph.D., Washington University School of Medicine

The Soft Bones Foundation research grant helped us complete DNA sequencing of the tissue non-specific alkaline phosphatase gene (TNSALP or ALPL) for our pediatric hypophosphatasia (HPP) patients at The St. Louis Shriners Hospital for Children. In 2015, we published our findings in a paper entitled "Hypophosphatasia: Validation and expansion of the clinical nosology for children from 25 years experience with 173 pediatric patients." Here, we reported our TNSALP mutation findings along with clinical details from our pediatric patients diagnosed with Odonto, Childhood, or Infantile HPP. This detailed analysis allowed us to refine the nosology for HPP and separate the Childhood form into "Mild Childhood" and "Severe Childhood" forms. Our findings should improve understanding of HPP presentation, natural history, complications, and prognosis. We continue to evaluate the role that TNSALP mutations play in women who are diagnosed with osteoporosis and treated with bisphosphonates, and subsequently exhibit an unusual fracture called "atypical subtrochanteric femoral fracture." These unusual fractures resemble those seen in Adult HPP. We proposed that some of these women may, in fact, have HPP instead of osteoporosis, and published the first case in 2012. We received a two-year grant from the National Institutes of Health to study this and reported initial findings at the 2013 American Society for Bone and Mineral Research meeting. We presented a second case of a woman diagnosed with "osteoporosis" showing a documented HPP mutation who suffered one of these unusual fractures. Our findings suggest that while TNSALP mutations may not have a major role in causing these unusual fractures, that it is important to distinguish the rare Adult HPP patients from the more common osteoporosis patient population. In 2016, I co-authored a paper entitled "Adult hypophosphatasia treated with teriparatide: Report of 2 patients and review of the literature." Here we reported mixed results for teriparatide therapy for HPP, but concluded that teriparatide shows some benefit for Adult HPP, which may be dependent on the number of TNSALP mutations (1 or 2) and the specific mutation(s) carried by the HPP patient

References:
Whyte MP, Zhang F, Wenkert D, McAlister WH, Mack KE, Benigno MC, Coburn SP, Wagy S, Griffin DM, Ericson KL, Mumm S. (2015) Hypophosphatasia: Validation and expansion of the clinical nosology for children from 25 years experience with 173 pediatric patients. Bone 75:229-39. Sutton RAL, Mumm S, Coburn SP, Erikson KL, Whyte MP. (2012) "Atypical femoral fractures" during bisphosphonate exposure in adult hypophosphatasia. Journal of Bone and Mineral Research 27:987-994. Sum M, Huskey M, Diemer K, Civitelli R, Gardner M, McAndrew C, Ricci W, Whyte MP, Mumm S. TNSALP mutation analysis in women with atypical femoral fractures and bisphosphonate therapy. J Bone Miner Res. 2013 28(Suppl 1): S40336.

Camacho PM, Mazhari AM, Wilczynski C, Kadanoff R, Mumm S, Whyte MP. Adult Hypophosphatasia treated with teriparatide: Report of two patients and review of the literature. Endocrine Practice 22: 941-50.

2015 Recipient - Luke Mortensen, Ph.D., The University of Georgia

Mesenchymal Stem Cells, or MSCs, are an adult stem cell type that produce bone, cartilage, and fat. These cells can be harvested from adults, and easily expanded to generate millions of cells from just a single donor. The potential to make new healthy bone has led to their study as a potential therapeutic for bone diseases like osteoporosis and osteogenesis imperfecta. The goal of MSC therapy is to inject cells from a healthy donor into a patient with impaired bone formation, with the goal of the transplanted cells traveling to the diseased bone sites and integrating into the tissue to form strong healthy bones. However, these MSC therapies have had limited success due to small proportions of the transplanted cells arriving at the target destination and limited survival of these transplanted cells. In hypophosphatasia (HPP), little has been reported on MSC therapy. With the support of the Soft Bones Foundation Maher Family Grant, the Mortensen lab has used our advanced microscopes to evaluate the potential of MSC therapy in a mouse juvenile HPP model. We track labeled MSCs to the bone marrow in mice with single cell resolution. We have investigated the effect of MSC therapy on the mortality and weight gain of severe HPP that is typically fatal by 3 weeks of age. We are currently working to develop high resolution imaging of living bone quality using a cutting edge technology called second harmonic generation to understand the effect of HPP on bone organization and the potential effects of MSCs and other therapeutics on this structure. Additionally, ongoing experiments aim to enhance the receptiveness of bone marrow to improve transplanted cell survival, and to engineer MSCs to enhance their homing and survival in the bone.

2016 Recipient - Brian Foster, Ph.D., The Ohio State University

I study the biology of tooth development, focusing on factors that are important for proper tooth formation and function. My emphasis is on the mineralized (hard) tissues of the tooth that give it mechanical strength- the enamel, dentin, cementum, as well as the bone that forms the socket around the tooth. By better understanding how teeth develop, I hope to gain new insights that may one day improve approaches for repair and regeneration of dental tissues.

Many people with HPP experience dental problems associated with the disease. A greater understanding of the effects of HPP on the bones and teeth has been gained from studies using a mouse model of HPP. This mouse model is a good representation of severe infantile HPP, however the severity of the disease prevents long-term studies, including those on therapies that may treat HPP-associated dental disease. As part of the Soft Bones research grant proposal, we aim to create new mouse models of HPP that vary in the severity of disease and location of tissues affected. These will include models that affect the bones and teeth, as well as those that primarily target the dental tissues. The novel HPP mouse models will then allow better controlled studies of how different therapies can prevent and repair HPP-associated skeletal and dental problems. These models will allow expansion of HPP research by additional researchers in the future, thus providing new resources to widen HPP-related research. With the support of the Soft Bones 2016 Research Grant, we have been able to analyze two new mouse models for dental disorders associated with HPP. The original mouse model of HPP that has been used for two decades has provided a wealth of information about all aspects of the disease, however, was limited by its severity that shortened its lifespan and restricted the types of studies that could be performed. These new HPP mouse models, developed in part with the grant, replicate the effects of HPP on bone and tooth development, but the later disease onset and targeted effects on hard tissues has translated into longer-lived HPP models that can be studied over a longer period to more advanced ages. One insight already provided by these novel models relates to the development of periodontal disease around teeth that have been affected by HPP. This work was recently published in the January, 2017 issue of the Journal of Dental Research, one of the top ranking scientific journals in the field of Dentistry, Oral Surgery & Medicine.

References:
Foster BL, Kuss P, Yadav M, Kolli T, Narisawa S, Lukashova L, Cory E, Sah RL, Somerman MJ, Millán JL. Conditional Alpl ablation phenocopies dental defects of hypophosphatasia. J Dent Res 96(1):81-91, 2017.

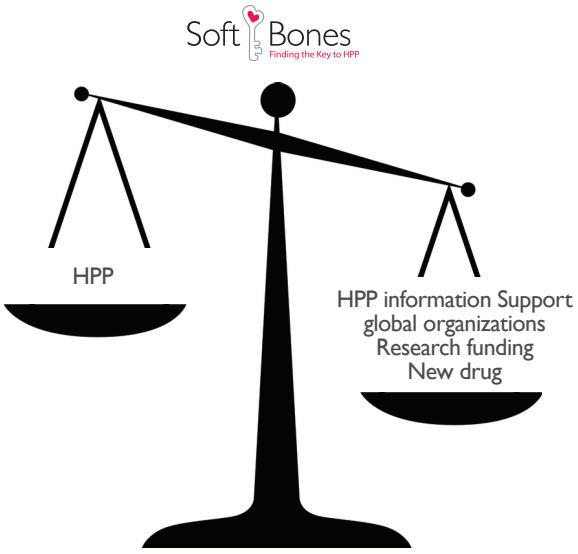
"LET'S CURE HPP"

BY GLORIA STONE, MBA, MS, G. STONE CONNECTIONS, LLC

Three words that mean everything to all of us. So much so you almost don't want to consider it because it seems such a lofty – even unachievable - goal. But ultimately that is what we wish for our loved ones and families.

But lofty goals and missions are what success stories are made of and we at Soft Bones are constantly assessing and realigning with the needs of the HPP community.

Before 2009 – the founding of Soft Bones – patients and families had few resources to better understand and manage their disease and there wasn't a community like there is today. Over the past 8 years, Soft Bones activities increased to align with new information on HPP, opportunities and needs.



Today, Soft Bones activities include:

- Jumpstart organizations in other countries to foster localized support and education
- Increase the scientific literacy associated with HPP through a Scientific Advisory Board
- Driving fundraising to award scientists' research grants to better understand the disease

Once Strensiq® (asfotase alfa) was approved by FDA – the first drug ever to be approved to treat HPP – on October 23, 2015, Soft Bones began to address issues associated with patient access to the drug. Once again, the organizational activities increased. As the HPP environment changes, so do the activities of Soft Bones.

More recently, the Soft Bones Board of Directors decided it was time to step back and more thoroughly align the needs of the HPP community to better understand the financial needs of the organization and the resources needed to meet the needs of the patient community. In 2013, the organization had completed a strategic planning exercise, and to the excitement of the board, the 5 year plan objectives were achieved in just over 3 years. So the board has voted to embark on another strategic planning session to revisit the mission and revise, if necessary, the Strategic Plan.

What is a strategic plan?

In the simplest of terms, it's a plan – a high level map to get the organization from point A to point B. The plan articulates the organizations priorities, the operations it will take to execute on those priorities and establishes a common goal. If effective, the plan will also not only articulate where the organization is going, but what actions will need to take place to make progress and to measure success.

We all want a cure for HPP and ultimately need to understand how Soft Bones can contribute to that.

The new plan is still in the works so there's nothing to share at this stage. However, the one thing we do know is that developing and implementing an updated Strategic Plan to support curing this disease will take the drive of all of the HPP community in whatever way people can contribute.

Now that there is a more robust rare disease environment out there, the next phase of Soft Bones in supporting a cure for this disease will likely require collaboration and more HPP information. We'll keep you posted, but this will be a plan for all of us to help drive a cure for HPP and to live our lives.

SOFT BONES AROUND THE WORLD

SOFT BONES UK

Soft Bones
United Kingdom

BY MERYL OCKENDEN

It is very exciting to have been asked to write about what we have been up to here in the UK. Looking back, it really has been a great few months and I'm happy to report, Soft Bones UK is off to a great start.

I will start by introducing myself, my name is Meryl and I was first introduced to HPP in 2014 when my son Maddox was diagnosed, at just a week old, with HPP. This is all still fairly new to me and we have had to learn quickly about this rare disease and about living life with HPP. I work in the healthcare profession as an Audiologist. I discovered quickly that working in healthcare and living with a child with a medical disorder are at other sides of the spectrum. Soft Bones US was a great support for me and my family whilst we were going through the motions and Maddox was extremely sick. It gave us hope and it was great to be able to connect with others that understood what we were going through. So last year through the NICE process and discussions with Soft Bones US we decided that a UK patient support group would definitely benefit us all and Soft Bones UK was formed.

December 2016 was a very busy month for Soft Bones UK. With the help of Soft Bones US we now have our very own logo featured above. We held our first charity ladies night in December which was a huge success and raised just under £3000, and we were involved in the first UK HPP paediatric clinic in Birmingham.

The clinic in Birmingham was full of children and families with patients as young as 6 months old to 12 years of age. It was so nice to see patients and families share stories and interact with each other and really comforting to be around people who understand the challenges HPP can bring to everyday life. The clinic was a huge success thanks to all the staff who put a lot of work into ensuring it ran smoothly and the HPP hippos went down extremely well with the kids. The next clinic will be in the summer 2017.

Looking forward, we have arrangements for a Soft Bones UK website. We are currently working on a date and venue for our first Soft Bones UK patient conference, which I am very excited about, so keep your eyes peeled for more information soon.

We are ever grateful for all the support we receive from Soft Bones US and the Brittle Bones Society and we are looking forward to the year ahead.

SOFT BONES CANADA

Soft Bones Canada

BY DEBBIE TAILLEFER

We had exceptional opportunities to be involved in a variety of activities and achievements. We inducted our Patient Advisory Board, who ran a highly successful contest to design and launch our new T-shirts. They did a fantastic job! We engaged various stakeholders to be a voice in health care, national and international rare disease groups, government and industry. SBC attended conferences such as the American Society of Bone and Mineral Research and Endocrine Society where we successfully engaged Canadian HCP's, scientists, researchers and students, endeavoring to raise awareness. We crafted Canadian tri-fold, genetics, patient information and physician awareness brochures which were placed in clinics and hospitals, sent out to patients for education about HPP and taken to appointments to shorten the wait time for appropriate diagnostics and to bring everyone up to speed on treatment. We developed our inaugural newsletter and designed a modern website, enabling us to reach out beyond our own walls and communities to keep Canadians up to date with HPP news and innovations and to reach new patients and families across the nation. A Café Scientific presentation in Montreal, PQ highlighted all aspects of HPP as a soft tooth and bone disease, and featured SBC's Scientific Advisory Board. In addition, they presented information about the drug discovery and the challenges of bringing a drug to market in Canada, especially for an ultra-rare disease. SBC was able to support bringing this presentation to TV on Canal Savoir and provide for sub-titles to help reach Francophone patients and HCP's. Definitely the highlight of our year had to be our Family and Professional Medical Conference held in July in Winnipeg, Manitoba. It was our desire to plan a very special occasion as well as ensure a well-rounded educational experience. We sought to have a variety of topics that provided a holistic perspective on how to manage and understand the disease, as well as hear about cutting edge research. Topics included an introduction to the science, as well as an extensive talk about the impact of the disease on the teeth. There was also exposition about how asfotase alfa operates in the body. A great deal of discussion was also generated about how the disease impacts individuals and families, both physically and psychologically. This rounds out some of our momentous goals accomplished in a very short period of time! We cannot adequately express what a privilege it was to be a part of reducing isolation and to see patients from all over Canada come together and be able to connect and spend time with each other. As difficult as it is that we share a rare disease, it is an honor to be associated with such a compassionate, courageous, resilient and inspiring group of people, as well as such a dedicated team of health professionals, scientists, advocacy and business leaders. You are definitely the reason we do what we do every single day!

FRIENDS FOR LIFE

BY TRACY WILLIAMS, MS

In this Creative Corner, Friends for Life Hippo Meets Mouse is dedicated to Dr. Jose Millan. After speaking with him, learning about his work and contribution to helping people with HPP, this friendly concept developed.

The day Soft Bones Hippo became friends with Miguel Mouse the universe sent a breath of fresh air to people with HPP and their families telling them about a new enzyme treatment that offers hope of a better life.

ADVOCACY IN ACTION

AT ASBMR – ATLANTA 2016

BY ZACH MCFALL 

During the Fall of 2016, I had the privilege of attending the 2016 American Society of Bone and Mineral Research (ASBMR) Conference. ASBMR is an organization that brings together clinical and research scientists involved in the study of bone and mineral metabolism as well as physicians from a wide range of specialties. ASBMR contributes to scientific research not only through annual conferences, but also through publications, advocacy and interactions with government agencies and other organizations. For conference attendees, there was always something to do. The exhibit hall housed all of the booths. These were occupied by drug manufacturers who discussed new drugs with physicians and researchers, companies who make medical devices for research, diagnosis and treatment, advocacy organizations like Soft Bones who spotlighted a disease or handful of diseases and an area devoted to researchers where they presented papers and posters of their current research. The other part of the conference consisted of lecture-style sessions for researchers, physicians and scientists. A presentation was given by the lecturer(s) and participants were able to ask questions.

As patient-advocates, we worked in shifts of a few hours at a time. We told our stories and answered questions of those who visited our booth. We also provided materials to physicians and scientists. One of the best give away items I've ever seen was a shiny metal key that doubled as a USB drive, which included materials from Soft Bones. This fit perfectly with the Soft Bones slogan "The Key to HPP." I really feel like we all made a contribution in connecting with and informing conference attendees. For me, the best part of the conference or any patient meeting is always being able to talk with and enjoy the company of other patients and/or caregivers. Because there are so few of us who have HPP, it is always a joy to talk and share with other patients. I also very much enjoyed sharing my story with scientists and physicians and answering their questions. As a science major, I very much appreciated the opportunity to attend my first scientific conference and be exposed to scientists and their ongoing research. Anytime you have the opportunity to take part in a Soft Bones event, I encourage you to do so. When we come together we share and learn in order to be more capable patients as well as advocates for ourselves, family and friends. Developing relationships with those who share your experiences and/or deeply care about and work so hard to help each patient with HPP, is very rewarding and fundamental in understanding the variety of ways HPP effects each patient.

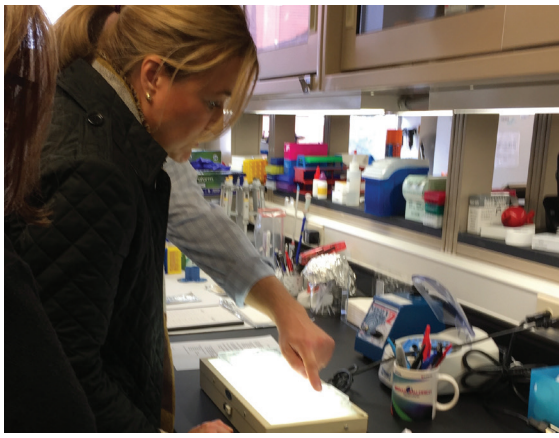
Most work done by Soft Bones, on a daily basis, is done without patient involvement, it's not flashy or put in the spotlight. These ladies help us find physicians, educate and inform us, advocate with insurance companies, governments and governmental agencies just to name a few. I cannot imagine how much work goes into all that they do to help us patients. I'd like to say a huge thank you to Deb, Denise, Adriane and Sue for all their contributions and hard work to fight HPP.

SOFT BONES
INAUGURAL HPP EXCHANGE MEETING AT OSU

BY SHARON TALKINGTON 

Today, we had the first Soft Bones HPP Exchange in Ohio. An Exchange is a new concept for a meeting where HPP patients, families, caregivers, doctors, dentists, researchers, and Soft Bones representatives all gather together to discuss HPP and related issues. This meeting was a dream come true for me and my family, as we've lived through doctors and medical physicians who did not know what to do with my husband's disorder. When we first started this journey we might hear of a doctor somewhere treating a patient with Hypophosphatasia (HPP), but we were never able to meet them face-to-face or schedule an appointment to speak with them. So this meeting was a shift towards more communication between patients, advocates, and medical staff as well bringing awareness to Hypophosphatasia.

The idea of this meeting began with a thought I had while I was attending Soft Bones Advocacy in Action at ASBMR this past September. While walking around Atlanta it occurred to me that we could possibly have a meeting at Ohio State University. I turned to Monica Baugh, who is from Ohio, and asked her, "Do you think we could have a meeting at Ohio State University?" She replied, "Sure, why not?" Then that Saturday night in Atlanta Soft Bones held a dinner for HPP patients, Alexion Pharma, and doctors who have contributed to HPP. Dr. Ing, an endocrinologist at Ohio State University, asked if we would like to do something similar to the event we were attending at OSU and if Monica, my daughter Rebekah, and myself would help organize it. We all happily agreed and I asked Denise Goodbar what we should do to get this meeting organized and planned. Denise grabbed Deborah Fowler and they spoke with Dr. Ing that night about doing an Info Exchange meeting. He already had a date in mind in November so there was not much time to get things organized. However, we all worked together through email and phone calls and by Nov 4, 2016 we were ready and the meeting was happening.



Top: In the OSU Dental research lab.

Bottom: Under the microscope a deer jaw from the archeology department.

Our dear family friend Dr. Rebecca Jackson (left) of Ohio State University Wexner Medical Center.

Dr. Ing helped us get a room at OSU and we all gathered there together. It was a myriad of HPP patients, caregivers, undiagnosed relatives, a genetic counselor, dental researcher (Dr. Brian Foster) and two of his associates, Soft Bones President Deborah Fowler and Denise Goodbar as well as Sue Krug and Dr. Luke Mortensen via video conference and in addition a lovely surprise visit from Dr. Rebecca Jackson. We enjoyed breakfast together, special goodies from Soft Bones, as well as a lunch that was delicious!

The information that we received during this meeting was so helpful to everyone gathered. First Dawn Allain, Genetic Counselor and Associate Professor of Clinical Medicine, Ohio State University, Wexner Medical Center, shared the path of how HPP can be diagnosed with genetic testing. She also talked about how we are still learning about HPP status such as carrier versus mild forms of HPP. Next, Dr. Foster, of Ohio State University College of Dentistry research, shared with us the basics of teeth development and structure as well as different things that can impact teeth development and dental health. He shared that the cementum is the most affected in an HPP patients teeth, but we need research in this area. He also emphasized more dental publishing about HPP in fields such as orthodontia is needed. It was interesting to hear him and his associates tell about their research in the lab and what they are working on. Dr. Foster has been working with HPP mice and is connected with Dr. Milan who works at Sandford Burhnam. They are currently working on ways to develop milder cases of HPP in the mice to see how they are affected by treatments. It's so exciting to see a growing interest in HPP with OSU's medical and dental faculty as well as Nationwide Children's Hospital.

In the afternoon, those who were able to stay, enjoyed a tour of Ohio State College of Dentistry with the Dean, Dr. Patrick M. Lloyd, as our guide. It was amazing to see the process students go through to become dentists and go into different specialties such as prosthodontics and orthodontia. At the end we had special clearance to go into Dr. Foster's lab to see the exciting work they are doing there. Research is a key and Soft Bones recognizes that every year with their grant. Last year Dr. Foster was the winner of the grant and we were honored to hear from him and see what he and his colleagues do in their lab.

Thanks OSU for hosting our first meeting in O-H... I-O!

Left: Dr Patrick M. Lloyd Dean of OSU College of Dentistry demonstrating first year student dental methods.

Right: Standing is Dr. Brian Foster winner of Soft Bones 2016 research grant in the OSU School of Dentistry research lab with staff and our tour group.

It is my belief that we are connecting the dots for HPP with the help of Soft Bones to move to a new level of treatment. Also, it is very likely, in my opinion, that Ohio will not only treat patients locally but also beyond state lines. I know in the past we would see specialists who had seen a patient(s) earlier that day who had come from out of state to see them and had been doing so for several years. Through hearing from others about their experiences with HPP, I know that many people travel out of state to start their HPP journey. However, I believe it is important that we connect with those that are close to home so as to have local support when a fracture occurs and have that relationship and continuity of care for HPP issues. Having both the support in far-away cities and close to home is a beautiful circumstance, and I pray that more people discover those around them rallying to their aid as my family has had people do in the past as well as in recent events in our lives.

This meeting gathered together doctors, researchers, patients, medical professionals, and caregivers to each have a voice in the discussion of what is going on with HPP, treatment, and what we can do in the future. I am so excited to have been a part of the process to facilitate the gathering of such a diverse group of people and I look forward to seeing more of the dots connected for an amazing, bright future with a cure for HPP.

IT'S A SMALL WORLD…

BY MONICA BAUGH

I work as a physician extender in a sports medicine office. Approximately four years ago our medical secretary asked if we treated hypophosphatasia. Surprised, I asked why as I was just going through my diagnosis and had not told anyone. She said there was a patient in the waiting room with a fracture and she had hypophosphatasia. I rushed to talk with the patient, Diane, and her husband. I told her I was very familiar with the condition, but our practice did not treat HPP. I explained that there was a physician at Ohio State University (OSU) who was familiar with the treatment of HPP, and our medical secretary, Terri, would assist her with getting an appointment. As she sat there with her husband, I could see that Diane was thoroughly disappointed. I could empathize with the fact that she had been told (like many of us "HPPers) that she couldn't be helped. Terri worked to get her a referral and a timely appointment with Dr. Ing at Ohio State. Because we were not involved in Diane's care I was unable to track her status; I only knew that the appointment was made. On November 4, 2016 at the Soft Bones HPP Exchange meeting at OSU, I finally got to meet Diane again and re-introduce myself as a fellow "HPPer." We talked about how her condition has improved and how happy we both were about our fortuitous meeting that day.

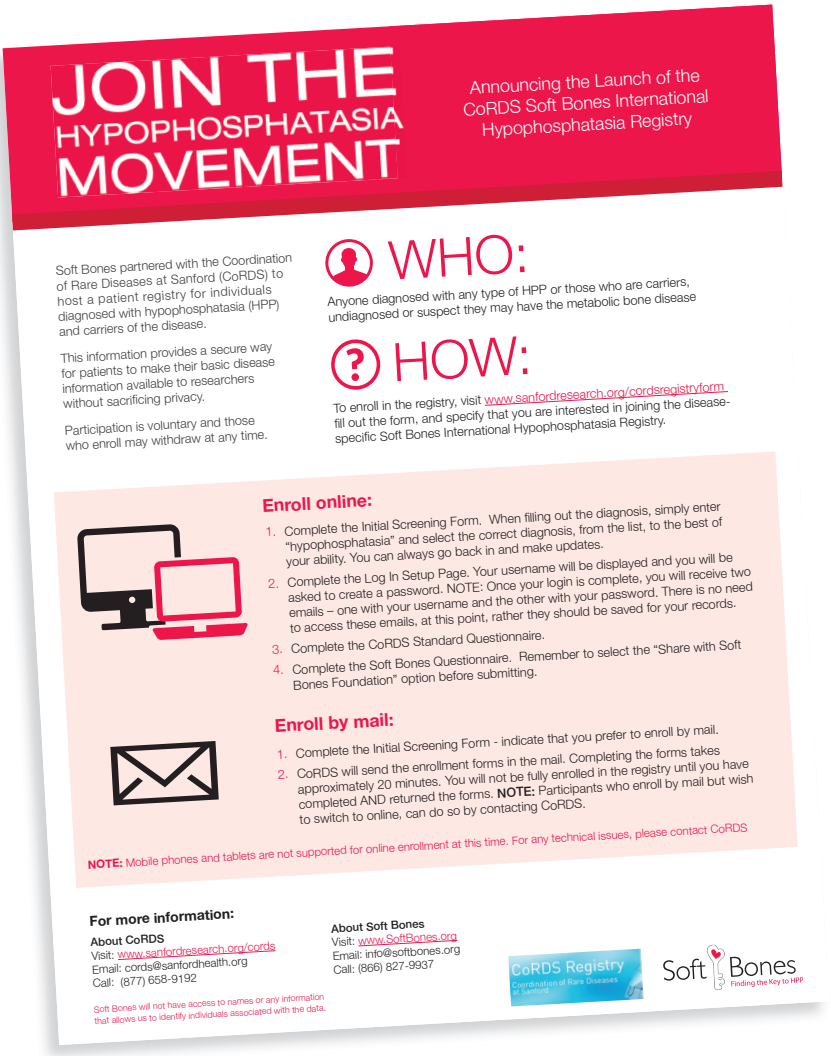
NEWS:

Soft Bones Emergency Cards Available: We have created two wallet-sized versions of the HPP Emergency Card for you to carry with you at all times. One version is a Strensiq™ Therapy Emergency Information Card (for those on therapy) and the other version is a General Information Emergency Card. You can download and print these cards out from our website http://www.softbones.org/resources/emergency-cards/. If you are unable to print these out yourself, please contact Adriane at adriane@softbones.org and she will be happy to send you one.

Annual Appeal: Thank you to all who donated to our first Soft Bones Annual Appeal. We raised just over $14,000.00. Your contributions help us to grow programs which increase awareness, offer support, provide education, and fund research for treatments and a cure. If you would like to make a donation, it's not too late. Please visit www.softbones.org or mail checks to Soft Bones at 121 Hawkins Place, #267, Boonton, NJ 07005.

HPP PATIENT REGISTRY

ENROLLS
100+
PATIENTS



JOIN THE HYPOPHOSPHATASIA MOVEMENT

Announcing the Launch of the CoRDS Soft Bones International Hypophosphatasia Registry

Soft Bones partnered with the Coordination of Rare Diseases at Sanford (CoRDS) to host a patient registry for individuals diagnosed with hypophosphatasia (HPP) and carriers of the disease.

This information provides a secure way for patients to make their basic disease information available to researchers without sacrificing privacy.

Participation is voluntary and those who enroll may withdraw at any time.

WHO:

Anyone diagnosed with any type of HPP or those who are carriers, undiagnosed or suspect they may have the metabolic bone disease

HOW:

To enroll in the registry, visit www.sanfordresearch.org/cordsregistryform fill out the form, and specify that you are interested in joining the disease-specific Soft Bones International Hypophosphatasia Registry.

Enroll online:

1. Complete the Initial Screening Form. When filling out the diagnosis, simply enter "hypophosphatasia" and select the correct diagnosis, from the list, to the best of your ability. You can always go back in and make updates.
2. Complete the Log In Setup Page. Your username will be displayed and you will be asked to create a password. NOTE: Once your login is complete, you will receive two emails – one with your username and the other with your password. There is no need to access these emails, at this point, rather they should be saved for your records.
3. Complete the CoRDS Standard Questionnaire.
4. Complete the Soft Bones Questionnaire. Remember to select the "Share with Soft Bones Foundation" option before submitting.

Enroll by mail:

1. Complete the Initial Screening Form - indicate that you prefer to enroll by mail.
2. CoRDS will send the enrollment forms in the mail. Completing the forms takes approximately 20 minutes. You will not be fully enrolled in the registry until you have completed AND returned the forms. **NOTE:** Participants who enroll by mail but wish to switch to online, can do so by contacting CoRDS.

NOTE: Mobile phones and tablets are not supported for online enrollment at this time. For any technical issues, please contact CoRDS

For more information:

About CoRDS
Visit: www.sanfordresearch.org/cords
Email: cords@sanfordhealth.org
Call: (877) 658-9192

About Soft Bones
Visit: www.SoftBones.org
Email: info@softbones.org
Call: (866) 827-9937

Soft Bones will not have access to names or any information that allows us to identify individuals associated with the data.

CoRDS Registry
Coordination of Rare Diseases at Sanford

Soft Bones
Finding the Key to HPP

Soft Bones is excited to announce that the HPP International Patient Registry has enrolled more than 100 patients and carriers. Thank you to all who have enrolled. Reaching this milestone in a rare disease state is noteworthy because it can provide important insights, ultimately accelerating research in HPP. In rare diseases, one of the biggest challenges is having a cohort of patients to access to better inform researchers. In July of 2016, Soft Bones partnered with the Coordination of Rare Diseases at Sanford (CoRDS) to host a patient registry for individuals diagnosed with (HPP) and carriers of the disease. The hope is that the CoRDS HPP Registry will provide information in a secure way for patients to make basic disease information available to researchers without sacrificing privacy.

Anyone diagnosed with any type of HPP or those who are carriers, diagnosed or suspect they may have the metabolic bone disease may participate in the registry. To enroll, visit http://www.sanfordresearch.org/cordsregistryform fill out the form, and specify that you are interested in joining the disease specific Soft Bones International Hypophosphatasia Registry. Participation is voluntary and those who enroll may withdraw any time. Soft Bones will not have any access to the names or any information that allows individuals associated with the data to be identified. For more information, contact Denise Goodbar at denise@softbones.org.

To enroll, visit http://www.sanfordresearch.org/cordsregistryform

SOFT BONES ON THE RUN

Amy Fiscus, husband Steve, daughter Makenna (age 11) and son Kellen (age 7). Kellen has mild HPP. The race was 5k and a children's run through the elementary school. It was the first race they all did together. Amy has run 2 half marathons in her Soft Bones gear and is prepping to run another 5 k in a week and a leg of a marathon relay the following day. Thank you to the Fiscus Family for spreading awareness!

RARE DISEASE DAY 2017

Being diagnosed with a rare disease is a defining moment for many families. For Rare Disease Day, this year, Soft Bones developed a campaign called "My Disease is Rare and so am I" to raise awareness as well as to provide hope to our patients and families and to show the world that we have HPP but the sky is the limit as far as our talents go. We had 67 people participate and each photo reinforced the message that we are not be defined by our disease. The campaign was very well received. We had nearly 400 visitors view the photos on our website – a record in itself! Thank you to all who participated. To view the photos, go to our website https://tinyurl.com/l8vf9cv.

Upcoming Events

May 25-28
Soft Bones exhibiting-American Association of Pediatric Dentists Meeting, Washington DC

July 14-15
Soft Bones Annual Meeting, Kansas City, MO

September 8 -11
Advocacy in Action – ASBMR, Denver, CO

September 25
Soft Bones Ninth Annual Golf Classic

Follow Us

To ensure we are saturating social media channels so that patients find us no matter where they are communicating, Soft Bones has launched two additional social media channels in addition to our Facebook page. You can now find us on Twitter and Instagram by searching @SoftBonesHPP. Please be sure to share with younger generations that are active on some of these other channels and see what we are up to around the world raising awareness of HPP!

Important Information For Patients

The Soft Bones Physician Referral Network

The physicians in this Network have given their consent to participate based on their interest in and experience with HPP. Other physicians may be added in the future. For a list of physicians in your area, via mail or email, contact Denise at denise@softbones.org or call 973-453-3093.

For more information, please contact the Soft Bones Foundation.

(866) 827-9937 – Toll Free
(973) 453-3093 – Direct Line
121 Hawkins Place, #267
Boonton, New Jersey 07005
www.softbones.org