

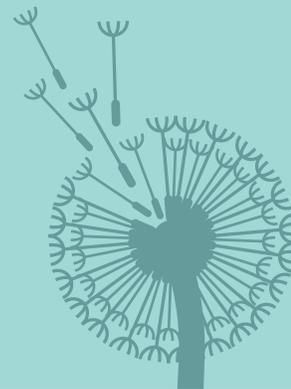
# Soft

2016 | Spring Edition



# Bones

Finding the Key to HPP



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## 7th Annual Soft Bones Golf Classic

This year marked Soft Bones' 7th Annual Golf Classic, thanks in part to an outpouring of support from donors, sponsors and participants. This year's event, hosted on September 14th at the Somerset Hills Country Club in Bernardsville, NJ, saw a record number 130 golfers. The Soft Bones Foundation remains committed to raising awareness of hypophosphatasia (HPP) and is thankful for the opportunity to come together with the local community at the annual Golf Classic.

The outing commenced with a delicious lunch before guests teed off, at noon, for a round of golf. The course featured two "Hole in One" challenges sponsored by Open Road Auto Group, Audi of Bridgewater and Audi of Mendham. The festivities continued into the evening with an extended cocktail hour reception, where guests could bid on a range of prizes in a silent and live auction. Some of the items up for grabs included tickets to a NY Giants football game, full orthodontic treatment for a child, and a weeklong stay at a beautiful home in Vermont. We were fortunate and grateful to receive an individual \$35,000 donation allowing us to offer our Maher Family Soft Bones Research Grant.



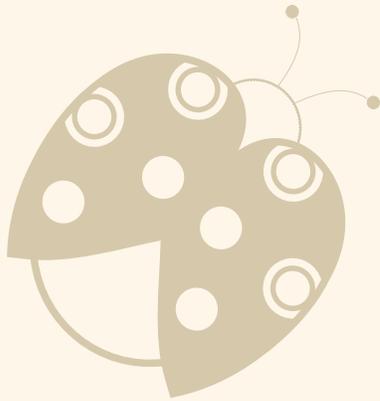
During the reception, event co-chair Bob Mulcahy, Soft Bones founder Deb Sittig and Roger Nettune thanked everyone for their generosity and highlighted the important work the Soft Bones Foundation has accomplished and their goals for the future. Deb's son Cannon, who has the childhood form of HPP, inspired the group with a few words on his journey with HPP and how he has never let his disease hold him back.

The annual Golf Classic is one of several fundraising events Soft Bones orchestrates to help raise funds and bring awareness to HPP. The proceeds garnered from the outing will aid in funding research and patient programs. A big thank you to those that joined — your support continues to help Soft Bones in the fight against HPP!



# Balloons For Kids

The Patient Advisory Board is proud to offer a new Soft Bones program called Balloons For Kids. As HPP families and caregivers, we know how difficult it is for kids to be stuck in the hospital and want to try to help lift their spirits. If your child has a surgery or procedure scheduled requiring a hospital stay, please let us know and we will send them a balloon. Contact [adriane@softbones.org](mailto:adriane@softbones.org) to arrange.



## New HPP Fact Sheets Under Development



The Patient Advisory Board (PAB) voted on topics for a variety of new fact sheets which will be published in the upcoming months. The PAB narrowed down the topics from a longer list and they include Dental, Disease Management and Nutrition. The Dental fact sheet provides an overview of what HPP patients should be concerned with when managing disease. The Disease Management fact sheet will address the multiple specialties that HPP patients should be considering in managing their disease. Finally, the Nutritional fact sheet will address some commondietary questions regarding HPP, including whether or not patients should take calcium and vitamin D.

Once approved, the Fact Sheets will be posted to our website under the Resources section at [www.SoftBones.org](http://www.SoftBones.org).



2016 —

# A Banner Year for Soft Bones

With a newly FDA approved treatment available to HPP patients, all eyes are on our community as we navigate the process of transitioning from trial to commercial drug. We are in the spotlight as physicians, healthcare providers and patients learn about the first ever treatment for hypophosphatasia. NOW is the time we need to be spreading the word, helping patients who could benefit from asfotase alfa see their doctors, get proper diagnosis and, if appropriate, treatment.

Part of our mission is to educate and one of the biggest challenges we have is a speedy and firm HPP diagnosis. Either patients are misdiagnosed or not diagnosed at all. Luckily, levels of alkaline phosphatase (ALP) in the blood is an easy sign for physicians to think HPP. But unfortunately, there are still plenty of physicians that overlook this simple indicator.

One of our goals this year for Soft Bones is to elevate the message: Low ALP – Think HPP. We are kicking off the campaign with a webinar which will be held in early March with a physician who will explain what ALP is, why it is important, what it does in the body and how it's related to HPP. We will also be working with labs and industry organizations to ensure that ALP is appropriately flagged on lab tests to alert physicians that the ALP is low and that further tests are needed to rule out HPP.

We are also pleased to provide patients with additional support with insurance and disability claims. The process can be time consuming and frustrating and we have been working with our partners to gather the right information to optimize a patient's chance of success. While we cannot guarantee that claims will be approved, we will be sure that providers are educated and have the appropriate information to make a decision.

Finally, the other big milestone for HPP this year will be the launch of the International HPP Patient Contact Registry. This registry will be the starting point for researchers, industry and physicians worldwide to pose questions to HPP patients to better inform care and research, hopefully leading to treatments and ultimately, a cure.

If you haven't visited our new website, please do. We are always open to feedback and of course, if you would like to share your story on our blog, please reach out to Adriane Eoga at [adriane@softbones.org](mailto:adriane@softbones.org).

Thank you and we at Soft Bones wish everyone a happy and healthy 2016!



**Please help Soft Bones by making a donation. No amount is too small!**

Go online to [www.softbones.org](http://www.softbones.org) to make a donation today. Your contribution is tax deductible.

# Alexion Uncommon Strength Campaign In Celebration of HPP Awareness Day

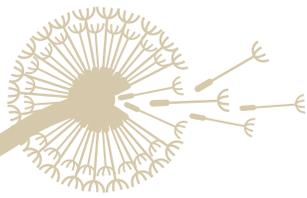


Alexion Pharmaceuticals unveiled their Global Uncommon Strength Campaign to coincide with the days leading up to HPP Awareness Day October 30th. The campaign centered around bringing the inner strengths and resilience of HPP patients and caregivers to the forefront by highlighting the unique attributes that help people in the HPP community persevere every day to lead an ordinary life. Components of the campaign included

- Campaign Website ([www.everydaywithhpp.com](http://www.everydaywithhpp.com))
- HPP Hero Illustrations
- Interactive Hero Builder

The Soft Bones Patient Advisory Board identified several patients and caregivers that demonstrated heroic qualities. These HPP hero illustrations were included as part of the initial campaign rollout.

The interactive hero builder allows users to build their own HPP Hero via an interactive online hero builder and share their hero creation with the community by submitting them, along with their story, to be displayed in the online gallery. To create your own hero illustration, simply go to the website [www.everydaywithhpp.com](http://www.everydaywithhpp.com).



## HPP Awareness Day — Up Up and Away!

To go along with the Uncommon Strength Campaign presented by Alexion, Soft Bones Foundation mailed out red capes emblazoned with the HPP Uncommon Strength logo to individuals who requested them. On HPP Awareness Day (October 30), people posted pictures of themselves, family members and friends wearing their capes to our Facebook page. In addition, Sue Krug created an HPP Hero frame which people used to post pictures in on that day as well. Thank you all for your enthusiastic participation!



# Soft Bones 2015 Patient Education Meeting in Orlando, Florida

On June 26 and 27, Soft Bones held its fifth Patient Education Meeting at the Disney Coronado Springs Resort in Orlando. Patients, caregivers, friends and professionals traveled from around the country to attend the meeting and gain valuable insight from leading healthcare professionals. Attendees also had the opportunity to meet others impacted by HPP, share information and form new friendships. The Soft Bones Foundation was able to offer a \$500 travel grant per family to help defray travel costs for those coming from a distance. Many chose to extend their stays to enjoy all that the Disney parks have to offer. The hotel even offered discount tickets and free transportation to the parks!

The event kicked off on Friday night with a meet and greet for all attendees. Deb Sittig, founder and president of Soft Bones welcomed everyone to Orlando. Deb also introduced all of the speakers. Refreshments were served and the room was filled with positive energy and conversation as people made connections. Many met for the first time while others were happy to catch up with old friends. It was amazing to see people who have been friends on Facebook for years meeting in person for the first time!

Saturday morning started off bright and early with a presentation by Deb who provided an update on Soft Bones activities and discussed the importance of advocacy for HPP. There are many opportunities for individuals to become involved in advocacy work. The louder and stronger our voices, the more we can accomplish together.

Dr. Matthew Drake, MD, PHD from the Mayo Clinic Endocrinology, Diabetes, Metabolism and Nutrition Cancer Center and a member of our Scientific Advisory Board spoke next providing an overview of

HPP. His talk, "Our Evolving Understanding of HPP", provided a thorough overview of HPP including symptoms, diagnosis and forms. He explained how HPP is more than soft bones and is a metabolic disease that impacts muscles, tissues and organs over time.

Next, Stephanie DeArme MHS, PA-C and Carol Elliott MHS, PA-C from the Division of Medical Genetics at Duke University presented "Hypophosphatasia: A Changing History". Their presentation focused on the genetic side of HPP including recessive and dominant genes and the difference between being a carrier and having the disease. They stressed the importance of having a geneticist and genetic counseling to go over family histories to try to determine the impact the disease may have on future generations.

The final presentation of the day, was given by Dr. Luke Mortensen from the University of Georgia. Dr. Mortensen is the latest recipient of our Maher Family Soft Bones Research Grant. Dr. Mortensen presented an overview of his research entitled "MSC Therapy for Soft Bones: Seeking Engraftment". Dr. Mortensen plans to use HPP mice to learn more about the therapeutic potential of transplanting alkaline phosphatase-rich cells into patients' bone marrow space. Dr. Mortensen's research premise is exciting and very promising and was referenced by the Soft Bones Scientific Advisory Board as a step towards a possible "cure" for HPP.

Thank you to all the patients, families, caregivers and professionals who were able to attend. Our close-knit community certainly enjoys being together. The information presented and the experiences shared were fantastic.



# Hip, Hip Hooray for Our New HPP Mascot The Hippo!



One of the accomplishments of our Soft Bones Patient Advisory Board (PAB) is the decision to create a mascot as a fun way to increase awareness of HPP in our communities. Working with Hypophosphatasie Europe, we asked for permission to use their current mascot, the hippo. Other animals were also considered and the PAB voted on the Soft Bones Hippo.

A special thanks to Steve Ursprung, President and Founder of HPP Europe, for presenting to his board and allowing us to share their adorable hippo concept!

The hippos will be part of our Rare Disease Day activities which will kick off in mid-February. We ask that patients that receive Soft Bones HPP Hippos take photos with them and post them to their social media profiles using the Global Genes profile picture frame overlay that will be shared in the following weeks. Please use the following hashtags [#RareDiseaseDay](#) [#WRDD2016](#) and [#SoftBonesHPP](#).



## Denise Goodbar and Adriane Eoga Join Soft Bones Staff

Denise Goodbar joined the Soft Bones organization as Program Manager in April of 2015. Denise is helping to drive forward the Soft Bones mission working with Deborah Sittig to implement the strategic plan set forth in 2013. Denise can be reached via email at [denise@softbones.org](mailto:denise@softbones.org).

Adriane Eoga is no stranger to the Soft Bones community. Many of you met her at the Orlando patient meeting as she helped with meeting set up

and childcare. In her new role, she will be responsible for welcoming patients, organizing patient meetings, coordinating volunteers and supporting patients with insurance and disability claims that are not covered by OneSource. She is also here to assist patients in a variety of ways including answering questions and providing them with physician referrals in their area. Adriane's email is [adriane@softbones.org](mailto:adriane@softbones.org).



# Soft Bones Launches New Website



October marked the launch of Soft Bones' new website, [www.SoftBones.org](http://www.SoftBones.org), which will serve as a hub for individuals living with hypophosphatasia (HPP), as well as their families and caregivers. The site features activities and resources relevant to the HPP community and delineates a comprehensive Soft Bones mission statement, with links to the Patient Advisory Board (PAB), Scientific Advisory Board (SAB), and Board of Directors. People looking for answers or information surrounding hypophosphatasia will have immediate access to materials through our sight. An online store is also in place allowing us to offer Soft Bones merchandise and to make donations to our community.

Through our Soft Bones blog, The Bone Zone, physician referral network and links to activist and fundraising opportunities, the new website will build a community for those affected by HPP and for those who want to help. In the upcoming weeks, an International HPP Patient Contact Registry will be launched where patients and caregivers will be able to register their information privately with the understanding that they will be contacted if patients are needed for a clinical trial or for research purposes.

The Soft Bones Foundation remains committed to increasing funding, awareness and necessary access for those with hypophosphatasia (HPP). The website will provide an important opportunity for individuals to connect, share, and take action around this disease. Visit us at [www.SoftBones.org](http://www.SoftBones.org).

It's Official —

## Soft Bones Canada

### Up and Running!

Soft Bones Canada has formally organized as a non-profit organization. Debbie Taillefer, Founder and President, led this heroic effort assisted by her Board of Directors including Lisa McGuffin and Jennifer Boin. Soft Bones US also participated in this process by sharing best practices and resources.

The Soft Bones Canada mission is to be a source of education, information, encouragement, and support for Canadian individuals and their families affected

by HPP, including interested individuals in the medical community. Look for their Facebook page Soft Bones Canada – Hypophosphatasia HPP Community and their website [www.softbonescanada.ca](http://www.softbonescanada.ca).

Soft Bones Canada is currently working on obtaining charitable status. For more information contact [debbie@softbonescanada.ca](mailto:debbie@softbonescanada.ca), [lisa@softbonescanada.ca](mailto:lisa@softbonescanada.ca) or [jen@softbonescanada.ca](mailto:jen@softbonescanada.ca).



## Soft Bones Awards

# \$25,000 Grant to

# Dr. Luke Mortensen

## to Promote Work in Hypophosphatasia



Soft Bones awarded its second annual Maher Family Grant to Dr. Luke Mortensen of Athens, Georgia based on his standout proposal for a new scientific approach to investigating this rare disease.



The Scientific Advisory Board for Soft Bones recommended Dr. Mortensen as the winner of the grant after thoroughly examining the worthy proposals of six applicants from the United States, Canada, and France, citing that a strong factor in their decision was his idea to use HPP mice to learn more about the therapeutic potential of transplanting alkaline phosphatase-rich cells into patients' bone marrow space.

"There have been challenges doing this for other bone diseases over the years, but Dr. Mortensen clearly understands the brief history of using marrow cell transplantation for infantile hypophosphatasia," said Michael P. Whyte, MD, Washington University. "We hope that seed grant support from Soft Bones might enable him to begin a long-term commitment for hypophosphatasia investigation."

Dr. Mortensen completed a post-doctoral fellowship at Harvard School of Dental Medicine and Massachusetts General Hospital before becoming an Assistant Professor of Regenerative Medicine and Engineering at The University of Georgia (UGA) in 2014.

Within UGA, Dr. Mortensen recently joined the Regenerative Bioscience Center (RBC). He works closely with the RBC director, Dr. Steven Stice, who is actively working with lab teams on a surgical gel, dubbed "fracture putty" that can heal a broken bone within days. Dr. Mortensen is in the process of opening his own research lab within the RBC, and hopes the seed grant money

will provide him the opportunity to gather preliminary data and develop the scientific justification for major external funding.

"I wanted to work towards something that can have an impact, and with HPP there is such a wide range of problems that patients can experience, yet so little research has been done," said Dr. Mortensen. "There is a great deal of potential to positively change people's lives with this project."

Dr. Mortensen formally accepted the grant in June at our HPP patient meeting in Orlando. This is second research grant Soft Bones has awarded since its founding in 2008. More information about hypophosphatasia and existing research can be found at [www.SoftBones.org](http://www.SoftBones.org).

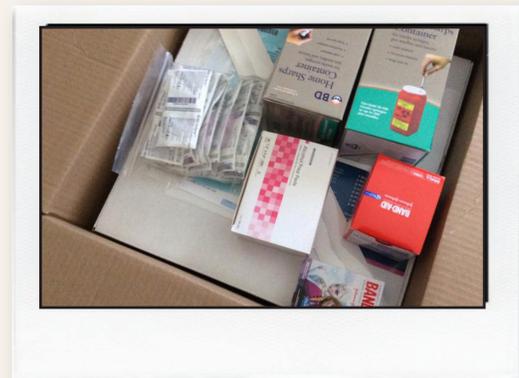


# First HPP Treatment Strensiq™ Approved

For the first time, the HPP community has an approved therapy. The U.S. Food and Drug Administration (FDA) approved Strensiq™ (asfotase alfa), in October, for the treatment of patients with perinatal-, infantile- and juvenile-onset hypophosphatasia (HPP). Strensiq, an innovative enzyme replacement therapy (ERT), is the first therapy approved in the U.S. for the treatment of patients with HPP. Strensiq is also approved in the European Union, Japan, and Canada.

“Asfotase alfa is an important advance for many patients with HPP, their families, and the medical community because it can effectively replace in the skeleton the deficient enzyme called tissue non-specific alkaline phosphatase,” said Michael Whyte, M.D., lead clinical trial investigator and Medical-Scientific Director of the Center for Metabolic Bone Disease and Molecular Research at Shriners Hospital for Children in St. Louis. “Without treatment, many newborns and infants with HPP fail to develop a normal rib cage and die from respiratory failure, and young children with HPP can suffer from rickets and muscle weakness.

Alexion offers support to patients with HPP through its OneSource™ program. OneSource provides each patient and family with personalized support from a dedicated Alexion nurse case manager, who can help patients understand their insurance benefits, receive reimbursement assistance, and provide education support such as in-home injection training. Patients, caregivers, and healthcare providers in the U.S. can now call **1-888-765-4747** to speak with a OneSource nurse case manager.



## Hats Off to HPP



Adding to our wardrobe collection of t-shirts, we have added knit caps to the list. The red knit caps are a cozy way to raise awareness of HPP this winter. The pom-poms add a touch of fashion flair too. The caps are for sale for \$25 each and help raise money for HPP. To order, visit [www.SoftBones.org](http://www.SoftBones.org) and click “Take Action” then products.

# Soft Bones Advocacy in Action in Seattle

The Soft Bones Foundation held an Advocacy in Action Weekend during the American Society for Bone and Mineral Research Meeting (ASBMR) in Seattle on October 9th – 11th. This was a departure from our typical Patient Meeting format in that each attendee had the opportunity to help staff our booth at the ASBMR annual meeting. This meeting brings together the field's most diverse and influential academic and clinical leaders. Patients were able to advocate for themselves and others directly with physicians, researchers and various medical professionals. A training session was conducted prior to the meeting to instruct patients on how to serve as effective advocates.

17 patients attended and each person volunteered a shift at the booth. The feedback received from the meeting attendees was overwhelmingly positive. Some researchers commented that they had never had the opportunity to speak with a person suffering from HPP and how they looked forward to returning to their labs to continue their work following this personal interaction. In turn, the patients loved the opportunity to share their personal experiences and in-depth knowledge with medical professionals.

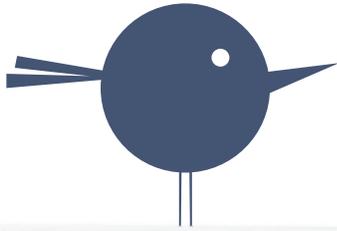
Soft Bones also sponsored a dinner on Saturday night attended by nine doctors/medical professionals, our patient advocates and their guests. In all 41 people enjoyed this very lively dinner. The evening offered an opportunity for individuals to share experiences, ask questions, and network with one another. One patient commented, "I used to feel alone, but I don't feel alone anymore".

On Sunday morning, attendees were able to watch a presentation made by Dr. Michael Whyte. The topic of the lecture was "Hypophosphatasia: The Journey to Treatment". This fascinating talk charted the progression of HPP from discovery up through the newly approved treatment. Dr. Whyte sits on our Scientific Advisory Board and is incredibly knowledgeable on HPP. Each person walked away feeling more informed.

The next Advocacy in Action meeting will be held at the Endocrine Society on March 31-April 2 in Boston.



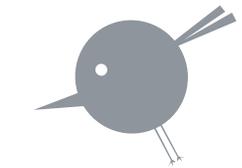
# Soft Bones Webinar



On April 27, 2015, Soft Bones presented a webinar entitled HPP: A Clinical Overview. Deb Sittig, Founder and President of Soft Bones US and Dr. Cheryl Rockman-Greenberg, MD, CM FRCP, FCCMG Program of Genetics and Metabolism, WRHA Professor, Depts. Of Pediatrics and Child Health and Biochemistry and Medical Genetics, University of Manitoba were our presenters.

Deb kicked things off with an introduction to the Soft Bones: The US Hypophosphatasia Foundation. Deb shared her own personal journey with her son, highlighted the Soft Bones organization, talked about what it means to be an advocate, and the importance of raising awareness. Dr. Greenberg gave a thorough presentation which included the history of HPP, the genetic components of HPP, the definition of HPP, the signs and symptoms of HPP, the types of HPP, and the diagnosis of HPP.

If you're interested in watching this webinar, go to our website [www.SoftBones.org](http://www.SoftBones.org) and look under the Resources tab. A big thank you to Dr. Greenberg for her participation and to Global Genes for coordinating the logistics.



## Mazel Tov to Shane Nachshen and a **Thank You from Soft Bones**

When Shane Nachshen of Mountain Lakes, New Jersey, was preparing for his bar mitzvah, he wanted to choose a mitzvah project that had personal meaning to him. He decided to become a certified referee so he could oversee youth soccer games and make money while doing it. Then, with the money he raised, make a donation to a charitable organization that was near to him. Shane decided to donate his earnings to Soft Bones to benefit families with HPP. During his bar mitzvah ceremony, he spoke of his dream of becoming an orthopedic surgeon, educated guests about HPP and why he chose Soft Bones as his mitzvah project.

Shane, on behalf of the entire HPP patient community, thank you for your kindness, generosity and for thinking of Soft Bones!





## Upcoming events



### FEBRUARY 29 |

Rare Disease Day.  
Please post pictures of  
you with your HiPPo



### MARCH |

ABC's of ALP Webinar,  
Rollout of International  
HPP Contact Registry



### APRIL 1-2 |

Advocacy in Action Meeting  
Boston, MA

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## Follow us on Instagram and Twitter

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!



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## 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 copy of the Soft Bones Referral Network via mail or email, contact Denise at [denise@softbones.org](mailto:denise@softbones.org) or call 973-453-3093.

### For more information, please contact the Soft Bones Foundation.

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