Soft Bones
2014 | Spring Edition
Finding the Key to HPP

Patient Education Meeting 2014

Soft Bones Rare Disease Day

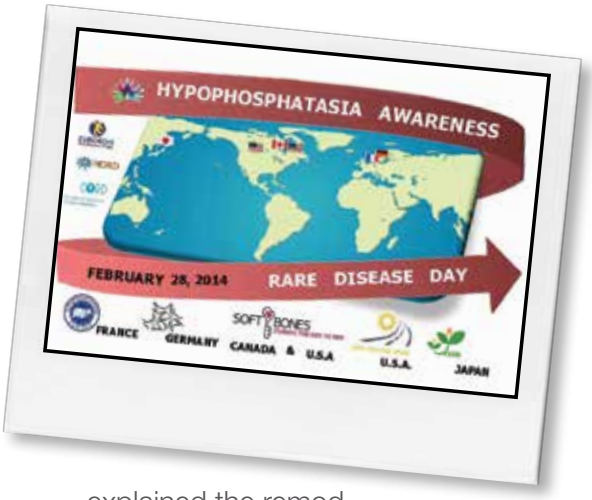
at Sanford Burnham, La Jolla, CA

As many of you may already know, February 28th is National Rare Disease Day! In honor of this, Soft Bones hosted its 2nd Patient Education Meeting at Sanford Burnham on February 27th - 28th. This conference was an opportunity for patients, families, caregivers and doctors to get together and discuss the disease while sharing personal hardships and accomplishments.

Although each patient is living with the same rare disease, their journeys are unique in many ways.

SoftBones founder, Deborah Sittig, opened the conference with an introduction welcoming all of the patients and doctors. Many patients explained how HPP affected their lives and their personal journey to getting their diagnosis, with Dr. Millan, Dr. Drake and Dr. Nunes listening intensely. Alexion Pharmaceuticals was also present, having one-on-one conversations with patients throughout the conference's meet-and-greet on day one.

Deb kicked off day two's events with an update on Soft Bones and their plans for a successful 2014, followed by Dr. Josè Luis Millan, a researcher at Sanford Burnham, with his presentation, "What Can Mice Teach Us About Our Bones?" Dr. Millan



explained the remodeling process; as we age, it takes the osteoblast longer to repair and fill in what the osteoclast has removed. His presentation was an in-depth explanation of how mice are bred to produce a "knockout mouse"—meaning researchers remove the entire gene of the disease, also known as TNSALP in HPP patients.

(continued on page 2)

(Patient Education Meeting 2014: Softbones Rare Disease Day continued)

He discussed how daily injections of ERT in mice caused the mice to grow bones and live longer. He stressed that although HPP research has come quite far, there is still much to learn about the disease and that feedback from the patient community is crucial.

Dr. Matthew Drake, a consultant in the Department of Medicine, Divisions of Endocrinology at the Mayo Clinic, spoke on "The Clinical Spectrum of HPP." Dr. Drake's presentation went in-depth on the varying types of HPP and some of the symptoms that dominate each certain type, explaining that there might be crossover symptoms.

Dr. Mark Nunes, a pediatrician, medical geneticist, and DNA diagnostician, rounded out the meeting with his presentation, "HPP: All in the Family." Dr. Nunes reviewed what HPP is and why it is difficult to predict how severe each patient's disease will be using the standard testing we currently have.

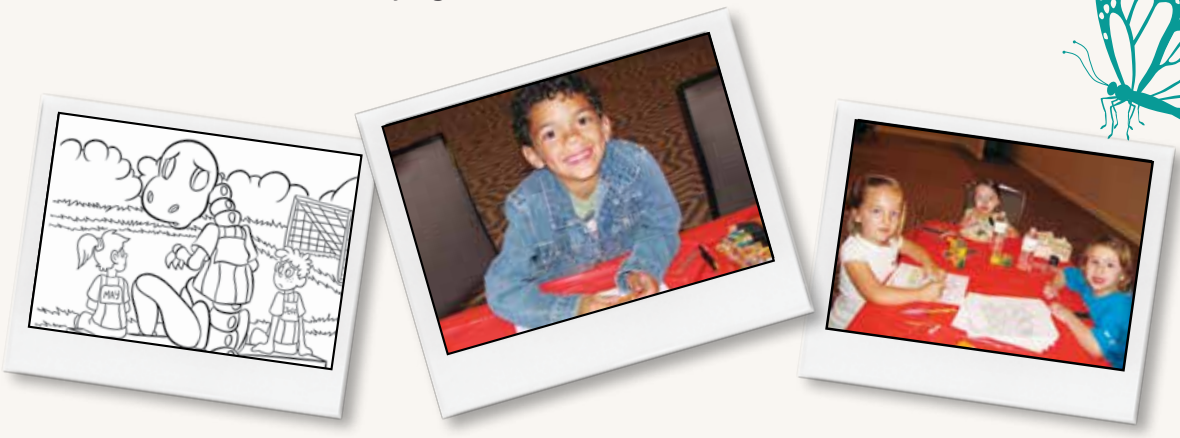
Using the "white sock, black sock" drawer theory, Dr. Nunes demonstrated a person's likelihood of having the mutated or normal TNSALP gene. He explained the genetics of HPP and how such genes can be dominant or recessive, demonstrating the probabilities of each type. He noted that differences could be found from other gene mutations not related to HPP which could either hinder or lessen the severity of the disease. Dr. Nunes emphasized that there is still much to be uncovered with DNA analysis. Dr. Nunes concluded by stressing that we are all in this detective game together!

The conference concluded with closing statements and helpful advice. The doctors were very informative and gracious in regard to answering questions and offering guidance. Overall, the conference was a great success, and a great way to appreciate National Rare Disease Day!

Coloring for a **Cause**

In order to bring the HPP patient community closer, Soft Bones and HPP Support Association of Japan collaborated to come up with a coloring contest. The contest includes a picture of the dinosaur from the HPP comic book that Jim Hawe created to help kids learn about the rare disease. The contest requires its contestants to create a name for the dinosaur in the picture, as well as color it in!

During the HPP International meeting, the HPP communities of France and Germany agreed to participate in the contest as well. All of the patient advocacy groups around the world really have been coming together to raise awareness about HPP. Everyone from patients to families and caregivers are participating in this contest to color for a cause, further raising awareness and educating many on HPP.



A Word from Soft Bones' Founder, **Deborah Sittig**

This is very exciting time for Soft Bones and our hypophosphatasia community, and I'm thrilled to be able to share some significant updates with you.

It's hard to believe but 2014 marks our five-year anniversary as an organization. We have accomplished a lot in a short time. As we have evolved, we built a strong foundation as an organization and at the end of 2013, we completed a five-year strategic plan to steer us down a path to fulfill our mission. The plan was extremely insightful and included recommendations on how to best serve patients while maintaining financial health as a not for profit organization. Our first step was to create a Patient Advisory Board to ensure we deliver the support and tools patients with HPP greatly need to keep them informed and make decisions about their care. A general Board of Directors will also be established.

We just had our Soft Bones Patient Summit, which took place at Sanford Burnham Medical Research Institute in LaJolla, California. It was incredibly inspiring to meet people from all over North America and hear their stories. At the meeting, we learned that Alexion is on track to submit an application for FDA approval for their first-in-class treatment for HPP, known as asfotase alfa, by the end of this year. We are all hopeful about the potential impact this drug can have on HPP patients.

One consistent theme that continues to emerge at our patient meetings is how the spectrum of disease varies from patient to patient. These insights always amaze everyone in the room, including the researchers and physicians who have studied HPP for years. This just proves how valuable it is to hear patient insights and to better ensure that all researchers and doctors have the information to better understand and treat HPP.

Finally, we are constantly hearing from patients and their families and friends asking about how they can help. One way patients can help is to share their personal stories about living with HPP with others. Hearing about your experiences and what you've learned empowers each and every one of us and can make a difference in someone's life. For families and friends, donations are the best way to show support. It is through these critical fundraising dollars that we are able to promote research, provide education for and about healthcare specialists and continue programs that support our community.

So spread the word about HPP and here's to a great 2014!

Please help Soft Bones by making a donation. No amount is too small!
Go online to www.softbones.org to make a donation today.Your contribution is tax deductible.

Alexion Strives to Increase HPP Disease Awareness Among Physicians

Many physicians are unfamiliar with diagnosing and treating ultra-rare diseases, such as Hypophosphatasia (HPP), which frequently leads to missed, delayed or inaccurate diagnoses.[1] According to global survey results published in the current issue of The Journal of Rare Disorders (JRD), patients living with rare diseases visited an average of 7.3 physicians before receiving an accurate diagnosis.[2] The survey also reveals that 44 percent of patients surveyed believed that a slow diagnosis resulted in delayed treatment with a negative impact on their condition.[2]

That is why Alexion is committed to raising HPP awareness and increasing the accurate diagnosis of HPP by physicians. Alexion recently launched a disease awareness campaign targeting a wide range of health-care practitioners, including endocrinologists, geneticists, orthopedic surgeons, rheumatologists, and nephrologists. The campaign includes increased presence at global, regional and local medical conferences and multiple print, electronic and interactive resources created to explain HPP and the impact it has on patients' lives.

"As a company, raising awareness about ultra-rare diseases is a critical component of our educational efforts as these patients often get lost or misdiagnosed with the more typical diseases that doctors treat," said Henric Bjarke, Vice President, Global Metabolic Disorders Franchise."Based on our experience treating other ultra-rare diseases, Alexion has focused the HPP campaign on highlighting the severity of this disease, and the importance of rapid and accurate diagnosis to help physicians recognize HPP in their practices."

As part of Alexion's expanded outreach programs to physicians at medical conferences, the company has attended a broad range of meetings, including the Endocrine Society's 95th Annual Meeting and Expo, the 63rd Annual Meeting of the American Society of Human Genetics, the American Society for Bone and Mineral Research (ASBMR) Annual Meeting and, most recently, the ACMG Annual Clinical Genetics Meeting. The ASBMR meeting is an excellent example of the diverse variety of physician specialties Alexion is reaching, including endocrinologists, geneticists, orthopedic surgeons, rheumatologists and nephrologists. ASBMR is comprised of approximately 4,000 members, including 1,600 clinical practitioners in attendance. These are the physicians that would most likely be involved in the care of a patient with HPP. These efforts point to the Alexion's commitment to physician education.

As part of the disease awareness program, Alexion has created a website for healthcare professionals. This website provides information to increase disease knowledge among physicians,and help them understand the importance of an early and accurate diagnosis of HPP.

On the website, physicians can find a clear overview of HPP, its pathology and its skeletal and multi-organ manifestations (ex. respiratory, neurologic, renal, rheumatologic, muscular) that affect patients. The website also highlights the danger of misdiagnosis – frequently occurring as its clinical presentation can overlap with other more common systemic diseases – and offers differentiating features of HPP, a solid diagnostic pathway to avoid misdiagnosis and an explanation of alkaline phosphatase (ALP, a vital enzyme) deficiency as the underlying cause of HPP.

The initial reaction from physicians around the globe regarding Alexion's HPP educational efforts has been positive as the company begins to help patients living with HPP receive a faster and more accurate diagnosis.

References

1. EURORDIS. The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe. 2009. http://www.eurordis.org/IMG/pdf/voice_12000_patients/EURORDISCARE_FULLBOOKr.pdf

2. Engel PA, Gabal S, Broback M, Boice N. Physician and patient perceptions regarding physician training in rare diseases: the need for stronger educational initiatives for physicians. J Rare Dis. 2013; 1(2):1-15.

Let's Outrun HPP

Cannon Ball Race

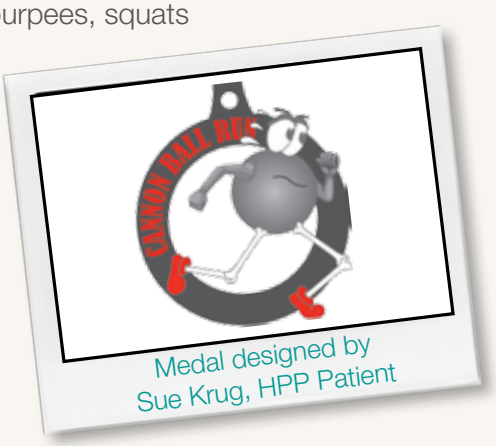
Join us on June 8th at the Cannon Ball Run to outrun HPP! The event is a unique way to challenge your overall fitness and support the fight against HPP. It's a two-part event like no other in North Jersey, where you will have the chance to meet Cannon Sittig, a local HPP patient and the name-sake of the "Cannon Ball Run."

Grab a partner and tackle the competition together, or approach it solo. On race day, registration will open at 7:00 AM if spots are available.

To guarantee you get a spot in the race, register online before June 1st. The Family 5k Race will begin at Birchwood Lake at 7:30 AM sharp, followed by the Full Competition at 7:40 AM. Pull your way out of the sand trap by completing seven minutes of three exercises: burpees, squats and push-ups! Then take on the challenging 5k-trail run through Tourne County Park.

The last day for registration is June 1st, unless maximum capacity is reached prior to this date. Registration gets you SWAG (Stuff We All Get) including a t-shirt, headband, goody bag, custom finishing medal and a donation to Soft Bones—along with some sore legs that you can brag to your friends about!

This race is no small feat. Feel great about your impressive accomplishment as well as your help in the fight against HPP! What are you waiting for – join us and let's outrun HPP! Go online to register at: register.bestrace.com



Medal designed by
Sue Krug, HPP Patient

An Update From

Dr. Whyte & Dr Mumm

Working with Dr. Nuria Gunanbens and her colleagues in Barcelona, Spain, Dr. Whyte and Dr. Mumm published an article in the April edition of the *Journal of Bone and Mineral Research.* The research described how some hypophosphatasia adults at the mild end of the clinical spectrum for that type of hypophosphatasia can present with "calcific periarthritis" – an unusual arthritis of especially large joints due to the seemingly paradoxical deposition of calcium and phosphate around joints yet without dental or skeletal manifestations of hypophosphatasia. Stay tuned for any further developments in Dr. Whyte and Dr. Mumm's hypophosphatasia research!

Rare Disease Day

State House Event

in Trenton, NJ

Rare New Jersey, a working group of patients, patient advocates and life sciences professionals working together to improve the lives of rare disease patients in New Jersey, presented an hour of learning and discussion, titled "Rare Diseases and the Life Cycle," on March 13th at the State House Annex Building in Trenton. The event was to celebrate Rare Disease Day and bring attention to all rare diseases to our state legislatures, pharmas, bios, physicians and others.

Co-sponsored by BioNJ, the state's trade association for biotech companies and affiliated organizations, and the National Organization for Rare Disorders (NORD), the event was held in honor of Rare Disease Day, an annual, global event that draws attention to rare diseases as an important public health issue that cannot be ignored.

Also on this day was the introduction of a resolution in New Jersey by Assemblyman Herb Conaway. Lisa Schill, a member of Rare New Jersey, and Debbie Hart, CEO of BioNJ, testified on our behalf to the Assembly Health and Senior Services Committee, and the bill was unanimously passed.

At the event in New Jersey, legislators, patients and the advocates who support them, highlighted the issues affecting rare disease patients in every phase of life. The event started with co-chairs Jane Castello from Soft Bones and Julie Raskin from CHI, welcoming everyone to the event.

Jane spoke about the wonderful support Rare New Jersey has been receiving from our state legislatures Senator Anthony Bucco of New Jersey's 25th legislative district and Assemblyman Herb Conaway from the 7th legislative district.

There were a total of eight patient and caregiver speakers at the State House event who discussed issues that include the diagnosis, the search for treatments and specialists, newborn screening and the affordability of care. Each speaker shared their personal stories. Working from the theme, "Rare Diseases and the Life Cycle," rare disease patients and their caregivers addressed how the impact of a rare disease begins even before birth. Rare disease patients face a myriad of obstacles in their quest for good health care and access to school, work, and housing. A luncheon immediately followed the program at the Lafayette Yard Hotel, which provided patients and caregivers with an opportunity to meet other members of the rare disease community and the people who support them.

The State House event served not only to celebrate Rare Disease Day and bring much needed attention to rare diseases throughout the state, but also as a space for rare disease patients, advocates and caregivers to discuss the many obstacles of living with a rare disease and get the recognition they deserve.

Rare Disease Day
Patient Posts

For Rare Disease Day 2014, I celebrated having HPP by baking “bone cookies” for my physical therapy office and for the nurses at two hospitals in our town. I would have loved to do more but one burnt batch led to some decision-making.

To spread the positivity, I created some Rare Disease Day stickers that read, “I love someone who is rare,” and handed them out to everyone who attended.

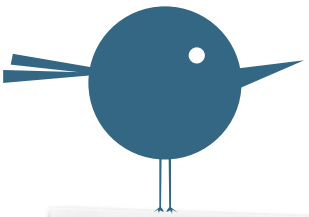
The best part of my day was sharing with everyone how awesome of a day it was. I added some glow in the dark paint to my boot as a way to commemorate the day with an encouraging statement: “It doesn’t take sticks and stones to break my bones but it takes more than adversity to break me.”

-Julia Beckley

To celebrate Rare Disease Day, my daughters, Evie and Lyla, took brochures to their classes and each did a short presentation on what Hypophosphatasia is. They explained that Evie has it and what Rare Disease Day is. They talked about how it’s important that people know about rare diseases so that others can get diagnosed. It was cute!

-Lindsey Elsaesser

HPP Europe Update



Sick and alone, global visibility of hypophosphatasia, a rare, genetic bone and dental disease was achieved with the first personal website created by Steve Upsprung. His objectives were to break down the isolation, find other patients, and to share and disseminate information.

MAY 2005 – MAY 2014

In May 2005, the first association in the world to help in the fight against hypophosphatasia was formed in France. It first focused on spreading information about hypophosphatasia in Europe, but soon spread internationally. They wanted to identify and support new patients. They also wanted to advance knowledge of the disease with doctors and scientists and to try to promote scientific and medical research.

TEN YEARS OF SHARING AND FIGHTING

The French network of experts

MEDICINE AND RESEARCH

Hypophoshatasia Europe has organized two international symposia in 2007 and 2012 bringing together global hypophosphatasia specialists to share their progress in their research. Several interesting therapeutic perspectives emerged, including Enzyme Replacement Therapy, which is progressing rapidly through clinical trials for very young children with a severe form of the disease. The association, thanks to the commitment of its members, volunteers and donors, has also been involved in financing many research projects aimed at better understanding the disease and its effects, fostering the conditions medical diagnostics, and to improve the medical care of patients.

COMMUNICATION

At the end of 2012, a book was published that contained information and explained terminology. It shows what the disease is and how the association is working with its partners. The English version of this book will be released sometime in 2014.

LOBBYING

During the past ten years, many lobbying activities were also conducted in collaboration with our partners, the Alliance Maladies Rares and Eurordis to encourage the recognition of rare diseases, help with the establishment of the “National Plans for Rare Diseases”, and help change the care of patients with rare diseases.

Another area of lobbying is pharmaceuticals, We were proactive within the industry working with companies willing to take an interest in hypophosphatasia!

The HPP Collaboration
Make Your Mark
Mosaic Murals

The HPP Collaboration: Make Your Mark Mosaic is a unique art project created by the HPP team at Alexion to partner with Soft Bones and the patients, caregivers and supporters of the HPP community. The project aims to build awareness around hypophosphatasia and inspire collaboration among people affected by HPP.

To create the murals, Soft Bones organized two events during HPP Awareness Week last fall, at Wildwood Elementary School, which Soft Bones founder Deb Sittig's son attends, and the Mountain Lakes Library. Each participant was given an 8-inch, square tile to decorate using blue and green coloring supplies.

"This is such a wonderful and fun idea to both bring the HPP community together and to educate the broader public about hypophosphatasia and the devastating effects it has on patients," said a third grade teacher at Wildwood Elementary School, Mountain Lakes, New Jersey. "The children and teachers at our event really enjoyed creating the tiles together to express themselves as individuals and to see the bigger picture of how together we can bring awareness of HPP."

The resulting tiles were then placed together to create two large maps of the globe, which are now 56 inches wide and 40 inches high!

To view The HPP Collaboration: Make Your Mark Mosaic mural, please visit **hypophosphatasia.com**. Everyone at Soft Bones and Alexion is hoping to see this project grow over time, so please continue to visit the website to stay updated on the status of this project and other HPP news.

Patient Feature
Susan Krug

Susan's health saga began at birth. Her parents were told that she would never walk or talk and would be mentally challenged. There were no treatments available and they were told to just love her every day because she wouldn't live past three years of age, because at the time no one with infantile HPP ever did. The doctors however, failed to mention anything about frequent bone breaks that would become a part of Susan's new normal.

By her first birthday, Susan weighed only nine pounds and could barely hold her head up, kick her legs or move very much. With HPP bones are extremely soft and brittle and can break easily. Challenges continued for Susan as fractures can happen even while standing up from sitting or getting a leg caught in the blankets at night. She experienced numerous fractures over the course of her life including many femur breaks leaving her in recovery for weeks or months at a time sometimes needing a wheelchair to get around. She also had to have numerous surgeries to put in rods when she had growth spurts to support her fragile leg bones. It got to a point where Susan's mom knew how to identify the breaks and how to splint them because fractures happened so often.

Susan never gave up, and neither did her mom; she was defying all odds against HPP. By the time high school rolled around Susan could walk on crutches and started to experience fewer broken bones. Despite not being like other kids her age physically, it didn't stop her from finding activities she was passionate about. She worked hard and lived life.

Because physicians didn't know a great deal about HPP since Susan's diagnosis, it wasn't until decades later that they would uncover she had two genes, including a novel form never seen in a living person, and the other form, infantile HPP.

Over the course of her life, Susan has experienced nearly 90 fractures, 29 rodding surgeries, 1 hip replacement, 4 level cervical fusions, dentures to replace fragile teeth and endless fractures of the vertebra.

Susan's case is indeed more severe than the average person with HPP. However, her iron-clad spirit despite life inside a "soft," fragile body never held her back.

HPP was previously unengaged with patient populations and families being dealt a difficult hand did not know what to do. There has been a significant paradigm shift thanks to others with the disease or caregivers with organizations being created such as Soft Bones: Finding the Key to HPP, helping to connect families in need, together encouraging for much-needed research, and finding necessary support and understanding.

Strength indeed comes in numbers and Susan is a strong voice in the HPP community and continues her quest to live and prosper, and to help others with the disease.

Congratulations to Sue Krug who was honored on May 8, 2014 by NORD and will be inducted to NORD's Portraits of Courage. NORD wrote in their announcement of Sue being selected for this honor, **"The committee believes that Sue embodies the spirit of courage and that her story will help send the message of hope and inspiration to others."**

Going Global

The 2014 Global HPP Patient Advocacy Group Meeting

Members from all over the world came together on February 15th to participate in this year's Global HPP Patient Advocacy Group Meeting in Zurich, Switzerland. Soft Bones, along with groups from France, Germany and Japan, met to discuss ways to continue our collaboration efforts. The groups updated one another on happenings within their country and shared ideas on how we can assist each other to help our HPP patient communities.

The groups met over lunch and were soon after joined by members from Alexion Pharmaceuticals. Together, the patient organizations and Alexion shared their upcoming plans and activities for this year's Rare Disease Day. The groups also discussed their accomplishments for the HPP community in 2013 and their plans for a successful 2014. The night allowed the global organizations to relax and enjoy each other's company at a local Swiss chalet, enjoying authentic Swiss cuisine including raclette and fondue.

Alexion kicked off day two with a full agenda. They started off with an update to the patient organizations on what they have been working on in 2013 and ended with the company's forthcoming plan for 2014. The participants were left feeling optimistic for the upcoming year, as all groups were able to learn how we can collaborate to help our HPP communities.

This year's meeting in Zurich proved once again the importance of the global community coming together to advance our understanding of this rare disease. The connections made globally have been invaluable in helping to keep the momentum strong in both research and in spirit.

Participants at this year's event included:

Gerald Brandt
Hypophosphatasie Deutschland

Steve Ursprung
Hypophosphatasie Europe

Hiroki Hara
Hypophosphatasia Support Association of Japan

Jim Hawe
Hypophosphatasia Support Association of Japan

Deborah Sittig
Soft Bones

Jane Castello
Soft Bones

David Heaps
HPP Choose Hope

Jennifer Heaps
HPP Choose Hope

Henric Bjarke
Alexion

Agnes Champigneulle
Alexion

Amanda Inalshigh
Alexion

Nyron Khan
Alexion

Roberta Mugnai
Alexion

Patricio Murias
Alexion

Gerrit Patterer
Alexion

Cahterine Payen-Champignois
Alexion

Mitali Rajan
Alexion

Upcoming events

JUNE 8 |
CANNON BALL RUN
MOUNTAIN LAKES, NJ

JULY 25-26 |
PATIENT EDUCATION
MEETING
PHILADELPHIA, PA

SEPTEMBER 8 |
THE 6TH ANNUAL
GOLF CLASSIC

SEPTEMBER 11 |
THE NATIONAL BONE
HEALTH ALLIANCE/RARE
BONE DISEASE PATIENT
NETWORK WORKSHOP

SEPTEMBER 12-15 |
ASBMR ANNUAL MEETING
HOUSTON, TX

OCTOBER 10-12 |
PATIENT EDUCATION
MEETING - ST. LOUIS

Important information for patients

The Soft Bones Physician Referral Network

The physicians in this Network have given their consent to participate based on their interset 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 Jane Castello at jane@softbones.org or call 973-453-3093.

For more information, please contact the Soft Bones Foundation.
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