HPS Network News

SUMMER 2015

From the Editor

Welcome to the HPS Network News! In an effort to increase the number of newsletters you receive and reduce costs, the print version of the HPS Network News will now contain just a sampling of news from the HPS community. It's exciting to have so much news, we can't fit it all in the newsletter! It see more news stories, visit: hpsnetwork.org

HPS Around the World: HPSers gather for the 22nd Annual Conference

Despite flight delays and cancellations caused by bad weather, a record number of people with HPS and their families, as well as HPS researchers, gathered in Uniondale, New York for the 22nd annual HPS Network Conference. In all, more than 280 attendees shared fellowship, information and education mixed with a healthy dose of fun.

The conference theme, HPS Around the World, was chosen to illustrate that while HPS is most common among people of Puerto Rican heritage, it does occur around the

HPSers race to create a costume during the getting-to-know-you session.

Kids attending the conference race to complete a scavenger hunt.

world.

The weekend was kicked off with some getting-to-know-you games, and for the first time this year, team building exercises. It will take the whole HPS team to find better treatments, and someday, the cure. Participants divided into teams and each team was given a bag of supplies to create an outfit suitable for a cruise around the world. Materials included scrap material, feathers, ribbon, cotton balls and a myriad of other craft products. After the outfits were created, they were modeled for the other attendees.

“I love the creative way the HPS Network is able to help us have fun, even though many of us are in pain or dealing with serious medical issues,” says Karen Tillman from North Carolina.

After dinner, Donna Appell, President and CEO of the HPS Network, presented her Year in Review outlining the work and achievements of the Network for the past year. During the Year in Review, a new research initative was announced – the Young Lung Centers.

Dr. Lisa Young, with the help of Dr. David Lederer, worked on a proposal for the HPS Network to join the Rare Lung Diseases Consortium. The Network is thrilled with the opportunity to strengthen our infrastructure and encourage our members to enroll in an HPS Center. This will enhance lung research and potentially enable us to be more organized should a drug trial be in our future. This opportunity was sponsored by the Office of Rare Diseases and the National Center for Advancing Translational

Teammates compete in the HPS version of CHOPPED!

Sciences. The announcement was received with great enthusiasm and several sessions at the conference explained the process. Dr. Lisa Young and Errine Garnett joined us at the conference and enrollment began on the second day. Twenty-seven individuals with HPS joined and the first of our Rare Lung Diseases Consortium HPS Centers became a reality.

Saturday morning began with our traditional opening with a song from Candice and Crystal Sipe, twins with HPS that make up the singing group, Angels in Voice.

"It is our way to be an inspiration," says Candice Sipe. Her sister Crystal added, "Maybe we don't always have the words to say to someone going through a tough time, but a song can be uplifting and comforting to them."

Next, the traditional family album circle allowed everyone to share what had happened to them in the last year, and what they were looking forward to in the coming year.

"I was blown away by how eve-

SS HPS visits Germany at the HPS Network Saturday dinner.

Nancy Lee and Julio Gonzallez dance together to celebrate Julio's lung transplant. Nancy had a lung transplant four years ago.

ryone seemed like family with each other and how they welcomed me with open arms. I left the conference feeling like I had gained a whole new gigantic family," says Eric Medina, who attended the conference for the first time.

After lunch, attendees were able to choose from a wide variety of sessions covering everything from HPS 101 for new families, to a panel of lung transplant recipients sharing their experiences, to a research focus group on the gastrointestinal disease of HPS. There were lots of fun sessions too!

One session involved a healthy snack-making contest modeled after the TV show CHOPPED. Other choices included Zumba lessons and yoga classes. Parents were able to attend a session with a panel of adults with HPS who agreed to answer ANYTHING that parents wondered about. Another session brought in the author of the book *Taking Your Child to the Doctor or Hospital*, Patty Weiner, to offer pointers about how to make interacting with the medical system less scary for kids.

While those with HPS and their families attended sessions, 28 researchers met for the second annual Meeting of the Minds to discuss issues related to HPS research and further collaboration. Dr. Donald Kohn, from the University of California Los Angeles, then gave an educational presentation about techniques involved with gene therapy.

Saturday evening the annual HPS Network Dinner set sail on a trip around the world. Themed as a cruise ship, attendees boarded via a gangway. Entertainment at the dinner involved several comical dances and skits at ports of call including: Germany, Ireland, Spain and Puerto Rico. Candice and Crystal Sipe, Ashley Appell and Casey Greer sang a variety of medleys to inspire the audience. This year they were joined by Yanelis Rivera Velez. Yanelis is a teen that was a finalist on the Spanish version of the popular show, *The Voice*. She gave the cruise stop in Puerto Rico an upbeat Puerto Rican flavor!

Sunday is traditionally science day. Researchers from around the country presented educational sessions about HPS, as well as new findings. Dr. Kohn also gave a talk, not so much about HPS in particular, but about how gene therapies in general are being developed and how they work.

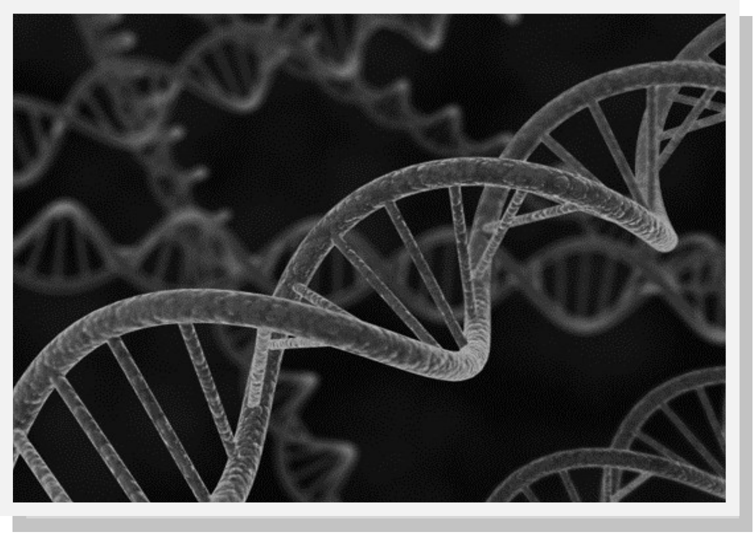
“The medical presentations on Sunday were the best ever,” says Nancy Lee, “It doesn’t matter how many years I’ve attended the conference, I learn something new every year.”

Once again, the conference was closed with a song from Candice and Crystal Sipe and a photo montage of pictures from the weekend. “I can’t put into words all the emotions I felt meeting my HPS family,” says Jessica Maldonado, a first-time attendee, “I learned so much. It was very uplifting, emotional and educational. It made me regret not being able to attend sooner. I’m already looking forward to next year.”

Are you interested in returning to NIH?

Are you are interested in returning to NIH to be a part of HPS research? Please let the HPS Network know. We are looking for anyone who was a part of the Pirfenidone study or anyone who has had significant changes in their health. Please contact the HPS Network at: 516 922-4022 or e-mail: info@hpsnetwork.org.

Meeting of the Minds explores gene therapy



The HPS researcher Meeting of the Minds was held, for the second time, at the HPS Network New York Conference in March. This researcher-only afternoon was attended by 28 scientists from around the United States.

HPS researchers presented updates on their work during the first part of the meeting. This collaborative forum gives HPS researchers a chance to share ongoing work with each other and discover areas where they can work together. Several of them later shared research developments on Science Sunday at the HPS Network Conference.

Dr. Donald Kohn, Professor in the Departments of Microbiology, Immunology & Molecular Genetics (M.I.M.G.) and Pediatrics and the Director of the Human Gene Medicine Program at UCLA, was the invited speaker. Although Dr. Kohn does not research HPS specifically, he is a well-respected expert in the field of gene therapy. He presented some of the latest research in the field of gene therapy to the HPS researchers. Discussion then ensued throughout the weekend among the researchers about how and whether these techniques could be applied to HPS.

“We were really excited that Dr. Kohn accepted our invitation and attended the conference. There is a lot of interest in the potential of gene therapy,” said Donna Appell, president and founder of the HPS Network, “It was wonderful to see science shared so willingly and so collegially.”

HPS Young Lung study is now open

HPSers interested in helping further HPS research may now enroll in the Young Lung HPS Research study. Initially, participants will be asked to sign consent forms giving the researchers access to medical records. They may also be asked to answer questions regarding their medical history over the phone. As the study continues, some participants may be asked to submit blood samples, pulmonary function tests, or CT scans. All participation is voluntary.

Although the study is primarily studying lung disease, the researchers are interested in those with any type of HPS from anywhere in the world. Identifying early signs of lung disease, even before symptoms ever appear, could help develop treatments, and someday even prevent, the lung disease of HPS. To do this, however, researchers must be able to distinguish between potential biochemical markers that all people with HPS share from those exhibited only by people with a gene type known to cause pulmonary fibrosis.

The research DOES NOT replace research being conducted at the National Institutes of Health. Rather, it is hoped it will help expand research as well as develop groundwork for future HPS studies and clinical trials. We hope this will offer an opportunity to participate in research for those not already being seen at NIH.

HPSers may participate if they are older than 12 and younger than 90.

For more information, or to learn how to participate, call: (516) 922-4022 or e-mail: info@hpsnetwork.org.

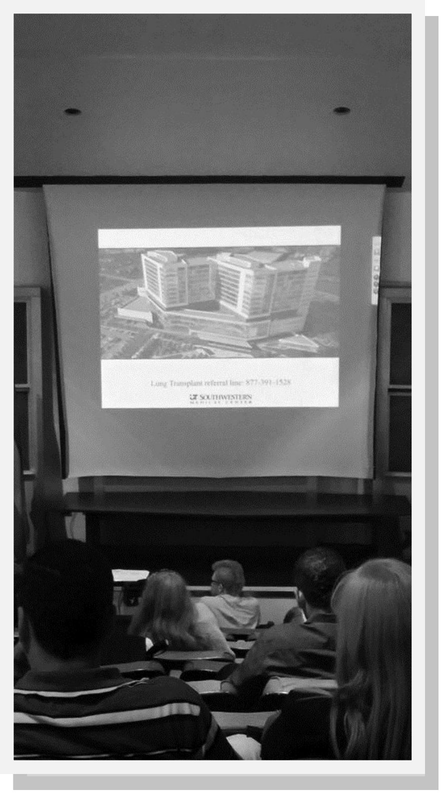
Visit the HPS Network at:

www.hpsnetwork.org

The HPS Network and the ATS Celebrate HPS Awareness

The HPS Network was honored the last week in March to celebrate HPS Awareness Week in Puerto Rico with the American Thoracic Society (ATS). ATS agreed to allow the HPS community to have our annual Lung Disease Week webinar live as well as an onsite event for patients at San Juan City Hospital.

HPS Network donors made it possible for the speakers to attend the event. The ATS provided the online broadcast, and lunch for attendees.



Attendees listen to a presentation on lung transplantation.

The Network received a wonderful reception from the families and physicians and had a spectacular event. San Juan City Hospital, part of Centro Medico Campus, was ready to greet us with a welcome banner in the lobby. Dr. Ricardo Fernandez, the Director of Pulmonary and Critical Care and Pulmonary Fellowship Training, was our host on site assisted by Dr. Maria Christina Betancourt. Their hospitality was incredible. Dr. Jose Hernan Martinez, the Director of the Hospital, met with our team and even gave a tour of the institution. Even Dr. Lidy Lopez, the Head of the Department of Health for San Juan, was in attendance.

Dr. Nieves Garrastegui, the President of the Pulmonary Society of Puerto Rico, gave a welcome address to all of the patients and doctors.

Courtney White from the American Thoracic Society was amazing. He saw to every detail to make this

Donna Appell, President and Founder of the HPS Network, addresses attendees.

meeting outstanding for attendees. He was so supportive and able to adapt to our specific needs seamlessly. The simulcast was managed and the room was filled with attendees that were engaged and interested.

Dr. Bernadette Gochuico, from Dr. William Gahl's team at the National Human Genome Research Institute of National Institutes of Health, Bethesda, Maryland, gave a lecture on the Pulmonary Fibrosis of HPS. She spoke about the Pirfenidone trial results and the lessons learned from individuals with pulmonary fibrosis from the past and the present.

Dr. Fernando Torres joined us from the University of Texas Southwestern Medical Center. He is the Director of the Lung Transplant Center and gave an informative talk about the preparation needed for lung transplantation in an HPS individual.

Dr. Yeidyly Vergne, hailing from the Ponce School of Medicine and Health Sciences, presented her research about the epidemiology of pulmonary fibrosis on the island of Puerto Rico.

The last session was entitled "Being Part of the Solution" and was presented by Carmen Camacho MA, LSW and Donna Appell RN. It summarized what each family needed to do to make informed decisions regarding lung health. Carmen felt that, "The ATS's efforts to educate our HPS community really created an impact, not only in patients but in the doctors and fellows that attended the event. Hearing about the possibility for lung transplants from the experts also created a sense that it is an option for people living in Puerto Rico, even though there is not a transplant center on the island."

This last session also included a discussion about the value of participating in research to help our community find a cure.

The NIH is actively involved in clinical research as they compile data regarding patients with the pulmonary fibrosis of HPS. Also the Rare Lung Disease Consortium has included HPS and centers are enrolling under the direction of Dr. Lisa Young from Vanderbilt University in Tennessee. Education about these protocols was provided by Donna Appell.

The HPS Network was both honored and grateful for the opportunities that the ATS provided to us. This onsite education event in Puerto Rico was an example of the ATS's commitment to "Innovations in Health Equality" and Courtney White was a perfect ambassador. The HPS Community in Puerto Rico was very pleased with the day and the efforts on their behalf.

"Having the ATS patient education day presented by experts in the field of pulmonary fibrosis and HPS gave us the opportunity to reach and educate many people in Puerto Rico and online.

Dr. Yeidyly Vergne, hailing from the Ponce School of Medicine and Health Sciences, presented her research.

It truly made a difference!" remarked Nancy Lee from the HPS Network Board of Directors. Nancy spoke onsite with the attendees and answered their questions first hand after receiving a double lung transplant four years ago.

Is your information up to date?

The HPS Network gets enquiries now and then from researchers and pharmaceutical companies that want to help us. They often want to know things such as:

- How many HPSers have a particular gene type?
- How many have the GI issues of HPS?
- How many HPSers are on the registry have pulmonary fibrosis?

The ability to answer these questions is important to the journey toward the cure.

Please remember to update your information with the HPS Network. Current contact information is critical to letting you know about research updates and opportunities. Also let us know if you learn your gene type (if you didn't know it when you registered). Let us know about any major health changes or upcoming surgeries.

Your assistance will help us be ready for the next advancement toward treatments for HPS.

The best way to let us know about updates to your HPS record is to e-mail: info@hpsnetwork.org. Include your phone number (even if we have it) to save time on follow up should there be any questions.

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HPS Parent Information Call

8:30 pm
English second Tuesday of the month
Spanish every Wednesday

Call: 1 (877) 394-5901
International or from Puerto Rico call: 1 (866) 999-7834

Enter code 7546770# (including the # sign)
For more information e-mail Carmen Camacho
ccamacho@hpsnetwork.org

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