

2023



# IMPACT REPORT

*Support that makes a real difference*



Everyone has a different story, a different journey that leads them here,  
but one thing remains the same,  
they found the support and guidance they needed from the **HPS Network**

Donna Appell, RN: Founder and Executive Director of the HPS Network reflects on how this all began:

"Terror is the only word I could use to describe the night that Ashley bled to shock from the bowel disease of Hermansky-Pudlak Syndrome. I kept thinking that she was going to die. She was in the ICU and struggling to return to that adorable two-year-old who was playing in her room earlier that night. The doctors told us they had not heard of the disease, and they never treated anyone with HPS. Those words were exploding in my head, competing with my urgent prayers to God for my daughter's health. It was a long hospitalization, and we were grateful to get her home. Immediately we thought about the next time and that feeling of mind-blowing isolation that filled every waking thought. We needed to find out if there were other families and if we found them, we wanted them to NEVER feel alone. We started the HPS Network 30 years ago and YOU have helped us accomplish more than we could ever imagine. What started with one family grew to help and support over 1100 families that continue to grow weekly. We provide seed funding to seminal research that has resulted in drug development. Our return on YOUR investment has proven time and again to be outstanding. Everything we do is to bring hope, fellowship, and a better quality of medical care to this community. We can and have delivered because of your generosity and kindness. It was your heart that kept ours from breaking beyond repair. You took away our isolation and we continue to show new families that they are not alone. Thank you for making such a difference in our lives and for investing in the work we do to help families live without terror."

*Donna Appell*

# A few HPS Network highlights

- Donna presented for multiple organizations including: The Milken Institute, The American Thoracic Society, Chan Zuckerberg Initiative
- HPS Leadership meets 3 to 4 times per week
- HPS Town Hall Meetings are held monthly to give updates to members and caregivers
- Peer Support Calls are held 5/month in both Spanish and English, facilitated by a social worker
- The Albinism International Databank (a patient reported survey resource) is under development
- Externally-Led Patient Focused Drug Development Report completed in English and Spanish and posted on our website
- Multidisciplinary HPS Clinics held every 3-4 months in Puerto Rico
  - Each Clinic averages 25-35 patients / 10-15 specialists in attendance



- 12 members participated in proof-of-concept research for an HPS bleeding treatment, which was successful and has now advanced to the next stage of research and was recently issued a patent
- Provide Observational Rotation for LIU Graduate Genetic Counseling Program (including weekly curriculum and thesis advisement)
- Continuously support research: our researchers have turned your funding for their initial research into RO1 awards from the NIH resulting in close to 8 million dollars of HPS research. Most of our investigators are still working on HPS.



- 2023 New York 3-day Family Conference
  - 200 attendees - including members, families, and medical professionals
  - "We're Drawn Together" - medical surveys and blood specimens for research collected on-site
- 2023 "Meeting of the Minds" - research workshop with 45 medical experts in attendance
- 2023 Puerto Rico Family Conference
  - 150 Attendees - including members, families, and medical professionals
  - Medical surveys for research collected on site
- HPS Puerto Rico Med/Ed Conference - Continuing Medical Education (CME) credits for 45 medical professionals in attendance
- American Thoracic Society (ATS) Collaborations
  - Annual Conference attendance includes HPS Educational Exhibit, Patient & Expert Forum, networking with pharmaceutical companies, and hosting of professional networking event: "Sundae on Monday"
  - Donna again became Chair of the Public Advisory Roundtable (PAR) in 2023 and a member of the ATS Board of Directors



## Ongoing and Looking ahead...

- Continuing to support HPS Gene Therapy at the NIH
- Funding another American Thoracic Society Research Grant
- Launching and loading the Albinism International Databank
- Developing an HPS Research Biobank
- Educating the community to be "Research Ready" for upcoming clinical trials
- Planning for 2024 HPS Family Conference in New York and Puerto Rico
- Increasing infrastructure in Puerto Rico to better serve the HPS population



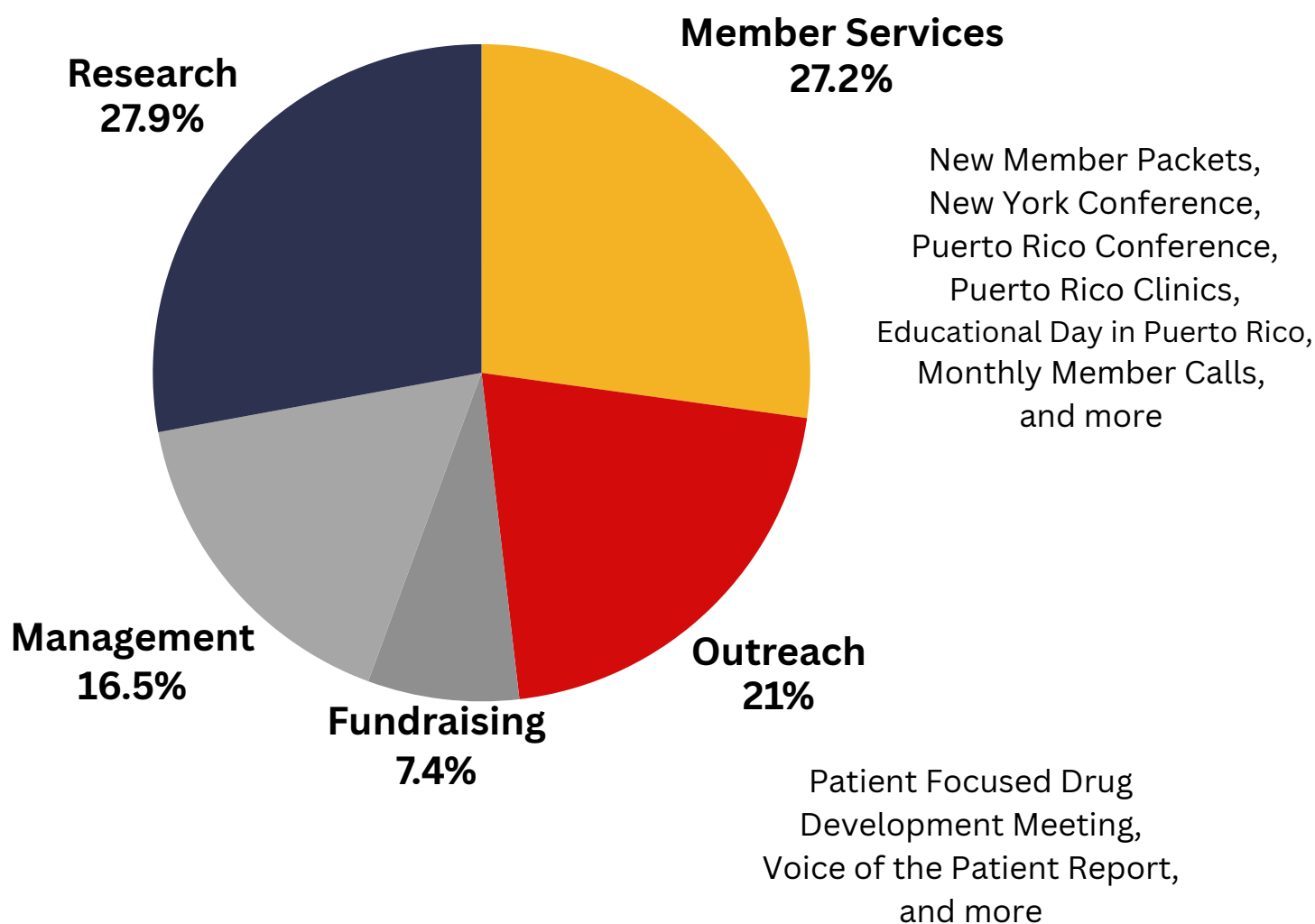


# FY 2022 AND 2023 FINANCIALS

<b>INCOME</b>	<b>Fiscal Year 2022</b>	<b>Fiscal Year 2023</b>
<b>Restricted</b>		
NOAH Grant \$	10,000.00	\$ -
Conference Fees \$	-	\$ 12,783.16
Conference Sponsors \$	-	\$ 11,527.80
CZI Grant \$	150,000.00	\$ -
EveryLife Foundation \$	5,000.00	\$ -
<b>Unrestricted</b>		
Giving Tuesday \$	4,681.39	\$ -
HPS Awareness Day \$	4,000.00	\$ 29,748.56
Lots of Dots Fundraiser \$	7,418.00	\$ 636.65
Christmas Concert Fundraiser \$	39,107.36	\$ 19,412.47
Annual Appeal \$	29,538.79	\$ 62,065.76
Local Fundraisers \$	4,005.57	\$ 2,945.36
Uncategorized Donations \$	171,970.73	\$ 153,534.95
<b>Total Income</b>	<b>\$ 425,721.84</b>	<b>\$ 292,654.71</b>
<b>EXPENSES</b>	<b>Fiscal Year 2022</b>	<b>Fiscal Year 2023</b>
<b>Programming</b>		
General Programming \$	81,567.06	\$ -
Member Services \$	60,531.02	\$ 182,931.63
Outreach \$	18,099.19	\$ 36,958.27
PFDD Meeting \$	86,498.19	\$ 32,105.75
Research \$	175,774.96	\$ 46,662.80
	<b>\$ 422,470.42</b>	<b>\$ 298,658.45</b>
Management & General	\$ 81,004.30	\$ 75,506.18
Fundraising	\$ 22,249.39	\$ 48,308.91
<b>Total Expense</b>	<b>\$ 525,724.11</b>	<b>\$ 422,473.54</b>
<b>Net Operating Income</b>	<b>\$ (100,002.27)</b>	<b>\$ (129,818.83)</b>
<b>Balance Sheet Assets</b>	<b>\$ 560,141.53</b>	<b>\$ 421,777.16</b>



## *Closer look at expenses*



# A few HPS Member spotlights



**Rick, Shelbie and Children**

"Getting our son's Hermansky-Pudlak Syndrome diagnosis in the Fall of 2018 was the most harrowing period of our lives. My husband was deployed, and I was figuring out motherhood alone. The guiding light through that period for us was undoubtedly the support from the ladies at the HPS Network.

Being able to pick up the phone and chat with the Network while I navigated uncharted territory as a new parent was invaluable. Because of the COVID-19 pandemic, we could not attend our first HPS Network Conference until March 2023. What An Experience! From conversing with other HPS families, meeting doctors, or the childcare provided for our young children, the Annual Conference was everything we could have asked for and more. We finally put faces to names of individuals who had been so helpful to our family for years. Leaving the conference, we felt very up-to-date on the medical research history and outlook for HPS because of the various seminars and learning sessions. The conference brought a level of understanding for us as a family that we didn't get from reading what is publicly available online. Sharing stories, issues, solutions, and ideas with other families provided the nuance and detail necessary for families living with a rare disease. We can't wait to get back in 2024."

"I began my journey with the HPS Network about 30 years ago when I was diagnosed with HPS 1. At that time there was very little information available until I found the HPS Network which became a valuable resource as I was planning my future. I have had an extensive, complicated bleeding history that has now affected my Panel Reactive Antibodies (PRA). As a result, finding platelets or a lung match will be very difficult, and a high PRA can disqualify you from been accepted to a lung transplant program. It took a lot of effort to convince my transplant center to take me! Now, due to my high PRA the wait will be long, and I might not even get my transplant, which is a thought that haunts me every day. I have been working with the team for over two years and I was finally listed December 14, 2021. I am very appreciative of that opportunity, but it did not come easy. It took a lot of commitment and convincing, which included my great team of doctors, my family and the support of my HPS family and the HPS Network. I realize I am blessed because not everyone has that experience. The HPS Network and I are committed to help every member receive that same support!! This is why I decided to work with the HPS Network and lead Patient support and education programs for those newly diagnosed or going through the transplant journey."



**Carmen**



**Cassandra and Casey**

"This February, we flew to Pennsylvania to participate in research at Cellphire. The bleeding disorder associated with HPS can be a difficult symptom to live with and it's very exciting to think that our blood samples will be used to help researchers as they try to develop new treatment options. We were given a tour of the lab and were encouraged to ask as many questions as we desired. As long distance friends, this was also a great opportunity for us to get to spend some time together. Between giving our samples, we got to explore the city. We enjoyed going to the book store, thrift shopping, and eating ice cream. The whole experience was a blast and we are glad we participated! We encourage everyone to take part in research if the opportunity arises. What we do today can make a lasting impact on future generations."

Stay up to date with the HPS Network and read new **"HPS Spotlight"** stories on our website!



# A few HPS Member spotlights



**Jill and Jeff**

"We are brother and sister, who both have HPS 5. For the first 50 years of our lives, we knew that we had ocular albinism, but after Jill had several complications from surgeries, her doctor suggested genetic testing for HPS. Jill began to search online and discovered the HPS Network. After reaching out to the Network, it was recommended that her brother should also be tested. We were given direction on the next steps for obtaining a diagnosis. It was so comforting to know that there was a community of others that dealt with the same physical challenges.

Each year the HPS family comes together for an annual conference in New York. We have both attended the conference, and have enjoyed learning more from the attending physicians along with others with HPS.

We have both made some lasting friendships.

The Network also provides monthly virtual meetings, where we can also connect with others in the HPS community. We learn about new research being done, and how to be our own healthcare advocates. We are also able to share life stories with our Network friends. We will always be grateful for the love and support of our HPS Network."

## **Dr. Enid Rivera, our expert Hematologist in Puerto Rico, and a member of our PR-HPS Research team, reflects on the establishment of the HPS clinics on the island.**

"Background: PR has the highest world incidence of HPS, which affects multiple health aspects. Many years ago, these patients were evaluated at different independent places, according to their acute health problem. Back in 2017 the first pediatric clinic took place at the University Pediatric Hospital at San Juan. The approach was completely different: longitudinal and multidisciplinary from 0 to 21 years of age. At the same time, an adult Pulmonary Fibrosis clinic began at Mayaguez. Eventually the adult clinic followed the pediatric multidisciplinary concept.

Purpose: Educate the patient and family to understand and accept HPS condition. Prevent and/or identify and address early health complications associated with HPS. Provide multiple services in the same visit, aimed at health promotion and maintenance. Reduce school and work absenteeism associated with multiple appointments and reduce costs and time for separate visits. To serve as an interdisciplinary management model for this population and for health professionals in training. How it works: The service revolves around the patient, with the various services: Hematology, Pulmonology, Pediatrics, Dental, and Vaccination. Samples are also taken to confirm HPS diagnosis using DNA analysis."



## **Dr. Samuel Seward is the HPS Network Scientific Advisory Board Chairman, a Board member and a longtime expert and friend of the HPS Network.**

"Within just a few months of completing my training, I had lunch with a special person—Donna Appell—who invited me to consider getting involved with the HPS community. That lunch sent me down a path to a wonderful career during which I have had the privilege of trying to make a difference in the lives of many, many people living with HPS. It is only through the generosity of our donors that the Network, and my work, continue to this day. I remain as committed as I was 20+ years ago to finding a cure for HPS!"



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