



2023

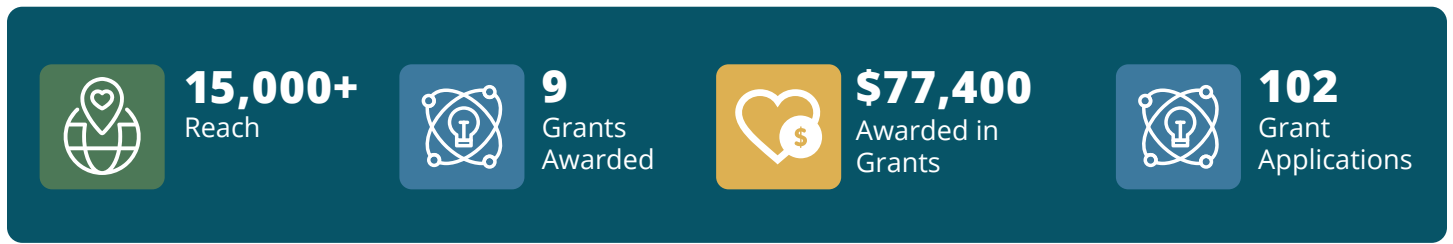
# Global Genes RARE Impact Grants Report





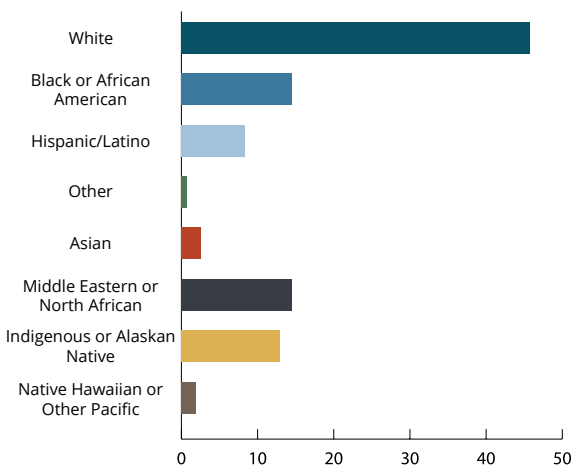


# Health Equity in RARE Impact Grant information

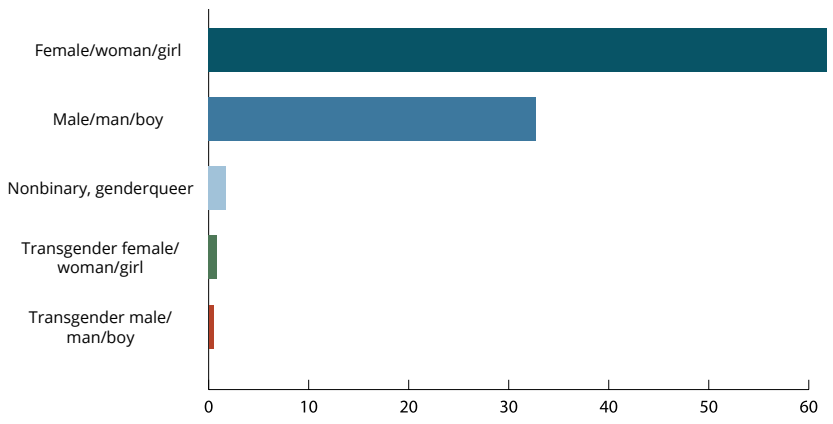


I never understood the importance of early screening and treatment until this Health equity project came to our village. Thanks to your efforts, my child's condition will be now manageable, and we are living with less fear and more hope.  
 - Kalumba Stephen, a caregiver and a Parent - Raising Hope International participant

Average Community's Race/ Ethnicity Composition



Average Community's Gender Composition







## Health Equity Grant Recipients

Nine Global Advocacy Alliance organizations received the 2023 Health Equity in RARE Impact Grant. The grants provided an opportunity for patient advocacy organizations to improve outreach strategies, develop content, and address challenges that affect underserved and underrepresented people within the rare disease patient community.

### Alliance to Cure Cavernous Malformation

While Alliance to Cure Cavernous Malformation has worked extensively to reduce health disparities in previous initiatives, this grant allowed the organization to adapt public-facing activities and materials to now include disabled patients. A group of Black, Hispanic, and disabled patients were also trained as patient ambassadors who will be available for media interviews and to speak at conferences. Furthermore, this grant has allowed the organization to update the website and social media to be more accessible and be WCAG 2.1 compliant.



### Hereditary Neuropathy Foundation

Hispanic populations often experience delays in diagnosis and barriers to treatment with Charcot-Marie-Tooth for a number of reasons. Hereditary Neuropathy Foundation launched a targeted bilingual awareness campaign in several NYC neighborhoods. The focus of the No Camines Solo (Don't Walk Alone) campaign was to educate the target population about CMT symptoms, diagnostic resources, and available and affordable treatment options. Diagnosed individuals and families learned about future clinical trials and genetic counseling services.



### Immune Deficiency Foundation

Immune Deficiency Foundation's Underserved and Undiagnosed program aimed to increase outreach efforts to improve diagnosis times and reduce barriers to care, specifically with Black and Hispanic patients. This was achieved through translation services, public awareness campaigns, and strategic partnerships.



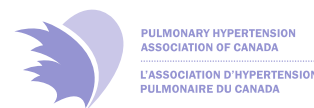
### NR2F1 Foundation

The Matrix platform, a patient-centered data collection platform, is currently translated into several languages, and through this grant, NR2F1 Foundation will now include ClinGen surveys in Portuguese, Hebrew and Korean. This work has expanded outreach into countries like Brazil, Angola and Mozambique where Portuguese is widely spoken, and South Korea (Korean) and the Middle East (Hebrew). NR2F1 partners with COMBINEDBrain to share translations with rare disorder umbrella groups (Global Genes/RARE-X, NORD, Eurodis) who can communicate the availability of the translated surveys to their member organizations.



## **Pulmonary Hypertension Association of Canada**

This project developed an Indigenous outreach strategy to provide Indigenous patients with culturally relevant resources to manage their disease and advocate for their care. Through this funding, PHA Canada hired an Indigenous Peer Facilitator to conduct an environmental scan to identify existing strategies and programs providing information resources to Indigenous people, key individuals who may be in a position to facilitate the outreach activities and best practices to guide the implementation. The Peer Facilitator also developed and conducted a focus group of Indigenous pulmonary hypertension patients to provide their insights and opinions. The focus group followed the National PH Community Conference. PHA Canada provided additional support to allow focus group participants to attend the educational sessions and connect with other pulmonary hypertension patients.



## **Raising Hope International Friends**

Those diagnosed with sickle cell disease face significant barriers to accessing care, and there is also a lack of awareness of the symptoms and causes which lead to delays in diagnosis. Raising Hope International Friends improved awareness and education, as well as open access to care and decreased the stigma associated with sickle cell disease in Uganda through the ministry of health, Village Health Teams.



## **The RUNx1 Research Program**

RUNx1 Research Program's En Beneficio de su Salud (For the Benefit of Your Health) provided educational and support resources for the Hispanic population. The program provided language-appropriate and culturally aligned materials to educate and create awareness around RUNX1 Familial Platelet Disorder (RUNX1-FPD), including learning about symptoms, potential transformation to cancer, and how to advocate for care. RRP also trained bilingual peer support advisors to serve as liaisons between the organization and the Spanish-speaking community who can facilitate peer support gatherings and lead outreach to patients.



## **Tanzania Sickle Cell Warriors Association**

With sickle cell disease diagnoses and mortality on the rise, Tanzania Sickle Cell Warriors Organisation sought to create education and awareness of the science behind the disease and how to manage it. Through education, the organization dispelled rumors and ineffective methods that the families have employed, and shared how proper medication and treatment can help reduce mortality of sickle cell disease in the region.



## **Team Telomere**

Through previous research, Team Telomere knows that the ethnic and racial make-up of those diagnosed with Telomere Biology Disorder (TBDs) is not representative of the population impacted by TBDs. Through this grant, Team Telomere sought to improve the capture of diverse clinical manifestations, barriers and health disparities in the TBD community. The organization conducted a community research survey using their Research Roadmap to better understand barriers to receiving care, geographic health disparities, accessibility of resources, equitable research representation, and diverse clinical manifestations.







For more information about supporting the RARE Impact Grants, contact [grants@globalgenes.org](mailto:grants@globalgenes.org). A special thank you to the sponsors of the 2023 Health Equity in RARE Impact Grant:





# RARE Mental Health Impact Grants Information



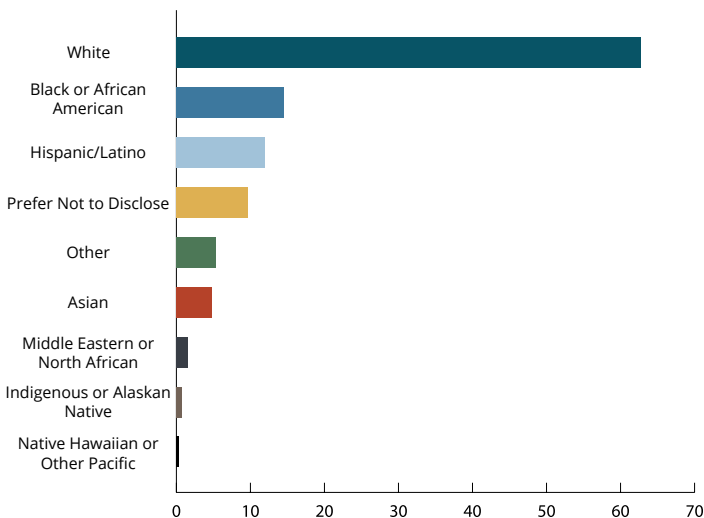
 <p><b>8,000+</b> Reach</p>	 <p><b>12</b> Grants Awarded</p>	 <p><b>\$169,822</b> Awarded in Grants</p>	 <p><b>130</b> Grant Applications</p>
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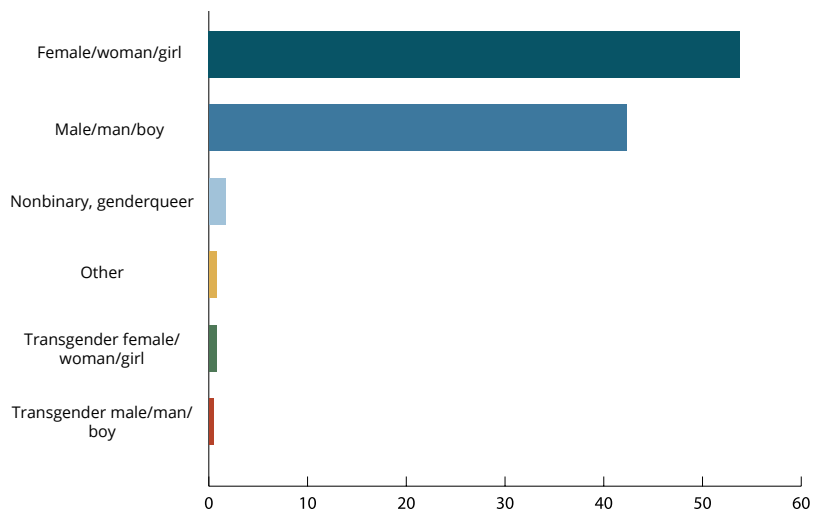
For the first time, we have been able to offer our families a much-needed professional mental health support program. We are excited to reflect on the outcomes of the grant as we plan our program moving forward.

- 2023 Mental Health Grant Recipient

Average Community's Race/  
Ethnicity Composition



Average Community's Gender  
Composition







# RARE Mental Health Impact Grants Recipients

In its inaugural year, the RARE Mental Health Impact Grants provided an opportunity for advocacy leaders to improve the health and mental well-being of the community groups they serve. Congratulations to the 12 organizations who received the 2023 RARE Mental Health Patient Impact Grants!

## **Angelman Syndrome Foundation**

Angelman Syndrome Foundation (ASF) began providing mental health support during the pandemic. Through this grant, the foundation expanded their mental health resources to high-need audiences who traditionally struggle to find effective support systems, including single parents, families living in rural areas, Spanish-speaking families, and those who have lost a loved one with Angelman Syndrome.



## **Beautiful You MRKH Foundation**

While the psychological challenges of living with MRKH are well-documented, adjustment to MRKH is an obstacle for parents and caregivers because few resources exist. According to qualitative data, parents experience self-blame, loneliness, guilt, and difficulty communicating with their children about MRKH. Through the RARE Mental Health Impact grant, the organization developed English and Spanish versions of a mental health toolkit for parents & caregivers, which was disseminated through webinars, MRKH family conferences and the Global Genes RARE Advocacy Summit.



## **The CUTE Syndrome Foundation**

The Cute Syndrome Foundation used this grant to expand its family support network to provide targeted 1-on-1 and group grief recovery support to adults, caregivers, and siblings experiencing anticipatory and bereavement grief. The support groups offered a safe, evidence-based environment for grievors to take effective and lasting action, no matter the type of loss experienced. Four volunteers were trained in The Grief Recovery Method, certified by the Grief Recovery Institute, and can now lead individual and/or group support sessions.



## **Dreamsickle Kids Foundation**

Individuals diagnosed with Sickle Cell Disease (SCD), and their family members, often experience mental health challenges, specifically depression. Depression can lead to a decline in physical health and require longer inpatient hospital visits, thus, increased medical bills, creating a brutal cycle. To address this, Dreamsickle Kids Foundation partnered with a local mental health agency to provide support groups and 1-on-1 therapeutic services to SCD patients and their caregivers. The mental health agency led support groups and provided uninsured individuals with discounted therapy services through their non-profit organization. By focusing on support groups and 1-on-1 services, Dreamsickle Kids was able to provide effective support to patients, parents, and caregivers, and gain a greater understanding of the concerns and anxieties SCD patients face.



## **ECD Global Alliance**

The Erdheim-Chester Disease Group Alliance (ECDGA) enhanced its virtual chat and webinar programs to provide social support, education, and mental health support to patients, caregivers, and grieving family members affected by ECD. The chat program included separate, regularly scheduled meetings for patients (monthly), caregivers, and those grieving the loss of a loved one (bi-monthly). Six webinars were offered, and recordings were made available to watch later.



## **Lymphangiomatosis & Gorham's Disease Alliance**

Lymphangiomatosis & Gorham's Disease Alliance offered a virtual mental health seminar open to patients over 16 years of age (with parental permission) who have a Complex Lymphatic Anomaly (CLA), their families, and/or caregivers. The seminar described common mental health issues when dealing with a rare disease, methods to find and communicate with therapists with experience in rare diseases and the importance of self care. It also helped the audience find a therapist or mental health service experienced with rare diseases. Patients and their families learned that their thoughts and feelings are valid and normal, and became more comfortable with the concept of self-care. Medical and mental health professionals were able to better identify, understand, and treat the needs of patients living with a CLA and their families.



## **The Marfan Foundation**

The Marfan Foundation used the RARE Mental Health Impact grant to integrate mental health into its annual conference in July 2023. This included an entire day of programming around mental health, with 30 workshops and sessions designed to raise awareness, educate, and provide tools and resources for people to take action. Experts in mental health and chronic health conditions led workshops and sessions on topics including coping with trauma and surgeries, stress and heart disease, and recognizing signs of mental health issues and getting help. Facilitated support groups were also held for family and caregivers around topics such as grief, loss, and caring for and loving someone living with Marfan.

THE **MARFAN**  
FOUNDATION





## **New England Hemophilia Alliance**

Individuals with bleeding disorders (BD) are often denied access to residential/inpatient substance use disorder (SUD) and mental health (MH) treatment facilities, despite being appropriate for admission. According to interviews with providers, people with BD are denied access for many reasons, including concerns about use of IV or injection medications, needles, fear about the medical complexity of BD, insurance issues, and access to medication. New England Hemophilia Association (NEHA) created educational tools and materials that provided community members with resources they can use to facilitate placement and what to do in the event of a denial. NEHA also raised awareness about the barriers individuals with BDs face when attempting to access inpatient/ residential MH/SUD treatment facilities, and encouraged community members to have conversations about MH/SUD issues with their BD providers.



## **Our Odyssey**

Our Odyssey serves young adults with rare diseases, ages 18-35, an age group disproportionately impacted by mental health issues such as anxiety, depression and isolation. The RARE Mental Health Impact Grant funded Epic Art, a virtual weekly program focused on therapeutic art-making to support mental health. Individuals who may struggle to talk about their mental health can access alternative means of communication, expression and connection through the arts. The program concluded with a virtual art exhibition of selected art pieces created during the program to spread awareness of the mental health concerns that can accompany a rare diagnosis.



## **Phelan-McDermid Syndrome Foundation**

Phelan-McDermid Syndrome Foundation (PMSF) recognizes that there has been an increase in the rate of devastating loss and medical challenges within the last few years. Through the RARE Mental Health Impact Grant, the organization launched a new mental health program to serve adults and caregivers of children with Phelan-McDermid Syndrome. The grant provided immediate mental health support for three specific caregiver groups, furnished a mental health professional who will facilitate a support group for these targeted caregiver groups, and helped to develop a strategy for a long-term mental health program that will become a part of our regular operations. The specific caregiver groups were: caregivers who have lost a child with PMS, caregivers whose child has or is experiencing neuropsychiatric episodes, and caregivers whose child is currently experiencing an extreme medical condition.



## **PTEN UK and Ireland Patient Group**

Individuals diagnosed with Phosphatase and tensin homolog (PTEN) and their caregiver are faced with many day-to-day challenges, from diagnosis, to cancer screenings, to the challenges of raising a child with autism or learning difficulties. The RARE Mental Health Impact Grant funded the administration of virtual support groups, training of facilitators, therapist presentations at the annual patient day event, and the creation of a survey that was issued to participants to assess the progress of the support groups. This program gave patients and caregivers an opportunity to speak to a trained professional, or each other, about the day-to-day challenges of living with this condition, or caring for someone diagnosed with PTEN.



## Usher Syndrome Coalition

The unpredictable and progressive nature of Usher Syndrome impacts an individual's identity across their lifespan, from being a hearing-sighted person to a person who is deaf-blind. Experts have highlighted the lack of therapists available to the deaf-blind community, which was only exacerbated by the pandemic, forcing an already isolated population, those experiencing hearing and/or vision loss, into further isolation. Facebook groups evolved as a way for adults diagnosed with Usher syndrome to provide peer support, but facilitators living with this condition are already dealing with their own cycles of loss and grief, and have not been trained in counseling, mental health, and life coaching. Through the RARE Mental Health Impact Grant, the organization offered a 10-week training series for peer group facilitators diagnosed with Usher syndrome, developed by a licensed psychotherapist living with Usher syndrome.

USHER SYNDROME  
COALITION

For more information about supporting the RARE Impact Grants, contact [grants@globalgenes.org](mailto:grants@globalgenes.org). A special thank you to the sponsors of the 2023 RARE Mental Health Impact Grant:









# RARE Meet-Ups Impact Grant 2023 Information







**900+**  
Reach



**6**  
Grants  
Awarded



**\$30,600**  
Awarded in  
Grants



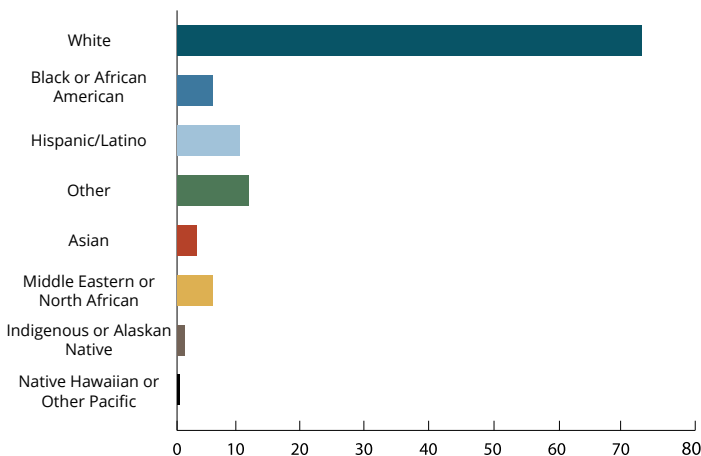
**129**  
Grant  
Applications



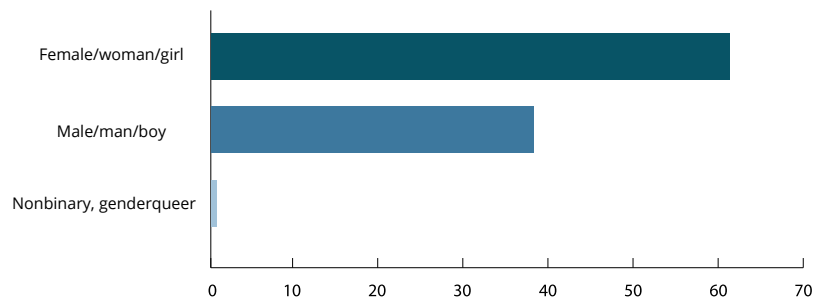
The biggest take away for me was that we all have a journey filled with appointments, doctors, specialists, individualized learning plans, etc. Our paths are all different, but we are in this together. For the first time in 6 years my husband and I didn't feel like our own tiny island.

- Coffin Siris parent

Average Community's Race/  
Ethnicity Composition



Average Community's Gender  
Composition





# RARE Meet-Ups Impact Grant 2023 Recipients

Six recipients received financial support in addition to marketing, materials, and resources for each Meet-Up. The Meet-Ups were held between June and October 2023, convening key stakeholders to foster collaboration, social support, education and awareness. As an exclusive funding opportunity for rare patient organizations that are a part of the Global Advocacy Alliance, this grant provides patient advocacy leaders with the opportunity to make a tangible difference in the lives of rare disease patients and caregivers across the globe.

## Charcot-Marie-Tooth Association

Angelman Syndrome Foundation (ASF) began providing mental health support during the pandemic. Through this grant, the foundation expanded their mental health resources to high-need audiences who traditionally struggle to find effective support systems, including single parents, families living in rural areas, Spanish-speaking families, and those who have lost a loved one with Angelman Syndrome.



## Coffin-Siris Syndrome Foundation

While the psychological challenges of living with MRKH are well-documented, adjustment to MRKH is an obstacle for parents and caregivers because few resources exist. According to qualitative data, parents experience self-blame, loneliness, guilt, and difficulty communicating with their children about MRKH. Through the RARE Mental Health Impact grant, the organization developed English and Spanish versions of a mental health toolkit for parents & caregivers, which was disseminated through webinars, MRKH family conferences and the Global Genes RARE Advocacy Summit.



## National Ataxia Foundation

The Cute Syndrome Foundation used this grant to expand its family support network to provide targeted 1-on-1 and group grief recovery support to adults, caregivers, and siblings experiencing anticipatory and bereavement grief. The support groups offered a safe, evidence-based environment for grievers to take effective and lasting action, no matter the type of loss experienced. Four volunteers were trained in The Grief Recovery Method, certified by the Grief Recovery Institute, and can now lead individual and/or group support sessions.





## **NEC Society**

The 2023 NEC Symposium, presented by the NEC Society in partnership with Cincinnati Children's Hospital, gathered hundreds from around the world to advance innovative research and quality care practices for babies affected by necrotizing enterocolitis (NEC). Those participating in this symposium as moderators, speakers and guests included patient advocacy groups and nonprofits, gastroenterologists, cardiologists, neonatologists, nephrologists, nurses, dietitians, patients and families. The 16 sessions during the symposium addressed novel research, collaboration, and advocacy.



## **Portuguese Association for CDG & CDG Allies**

Portuguese Association for CDG & CDG Allies provided both an in-person and virtual offering for those affected by Congenital Disorders of Glycosylation (CDG), which includes around 170 ultra-rare metabolic disorders. The in-person event was split into two events in July 2023. "First World CDG Advocacy, Policy, and Leadership Academy" was focused on capacity building for PAG leaders, advocates and family members seeking to improve advocacy skills, and the "6th World Conference on CDG" will appeal to the CDG community at large, including drug development, therapeutic approaches, current research and care guidelines. The virtual option, "World Think Metabolic, Think GDG Academy," included expert-led pre-recorded video sessions through the WorldCDG website. These sessions educated viewers on advances in CDG research and the drug development process, everyday CDG care and management, and encouraged attendance at the 6th World Conference on CDG.



## **Yellow Brick Road Project**

Yellow Brick Road Project will hosted its annual scientific and family conference July 31 - August 2, 2023. The 2023 HNRNPH2 Conference & Family Meeting gathered patients and families with scientists and clinicians who discussed the latest research, conducted patient evaluations for the natural history study, and provided patients with the opportunity to have evaluations on motor/PT assessments, neurological assessments, and EEGs. In addition, bloodwork and controls to create stem cells, and saliva swabs were collected to investigate X-inactivation/skewing. Skin biopsies were taken to cultivate patient-derived iPSCs. With less than 150 individuals diagnosed with HNRNPH2 related disorders, this in-person event was critical for helping the organization gather patient data and continue the National History Study, as well as allowed patients and families to connect to the scientific community and experience a sense of belonging.



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