ANNUAL REPORT

2025

Rosenau Family
Research Foundation

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Foundation Updates

From the Desk of the Executive Director

Gabriel M. Cohn, MD, MBA
Executive Director

As we close 2025—and mark a quarter century of progress in the 21st century—we recognize extraordinary advances in genetics, genomics and multi-omics, disease understanding, diagnostics, newborn screening, and disease-modifying therapeutics. For some rare diseases, these breakthroughs have been transformative. Cystic Fibrosis, once limited in therapeutic options and outcomes, now benefits from an expanding array of treatments that meaningfully improve quality of life and extend life expectancy. Yet for other conditions, including Krabbe disease, effective and accessible therapies remain elusive despite deepening insight into the molecular biology and pathophysiology that drive disease.

This Annual Report reflects the continued commitment of Rosenau Family Research Foundation (RFRF) to improving the lives of individuals and families impacted by Krabbe disease and Cystic Fibrosis through research funding and advocacy. Inside, you will find highlights from the publications and presentations arising from RFRF-funded projects, as well as seven newly approved, highly innovative grants—four focused on Krabbe disease, two on Cystic Fibrosis, and one to enhance AAV vector biodistribution to strengthen gene therapy approaches. Together, these publications and grants aim to advance foundational discovery, support translation to human application, accelerate new therapeutic strategies, and strengthen clinical knowledge and practice.

Because important gaps and challenges persist, we remain steadfast and forward-looking, focusing our efforts on the most urgent needs. The 2025 Krabbe Translational Research Network (KTRN) meeting asked attendees to identify the most critical gaps in Krabbe disease; the meeting's output—summarized in this report—should serve as a compass for the Foundation and the broader Krabbe disease community. Among the priorities identified was the need to cultivate newer, young investigators and clinicians, and we are pleased to highlight RFRF's initial efforts in 2025 to address that need.

Progress depends on partnership. No single person or organization can do all that is required to support these communities, and RFRF is proud to sponsor and recognize the organizations that advocate for and stand beside families and individuals affected by Krabbe disease and Cystic Fibrosis. We also extend our sincere gratitude to the researchers and clinicians advancing the science, to our dedicated staff who carry out our mission every day, and to the members of our Board of Directors and Scientific Advisory Committee, whose leadership and guidance strengthen our work. Thank you to those who have served, and to those who have recently joined us, as we continue this critical, challenging, and most noble of journeys.

Gabriel M. Cohn, MD, MBA

Our VISION

Living a life undefined by Krabbe disease and Cystic Fibrosis

Our MISSION

Improving the lives of people impacted by Krabbe disease and Cystic Fibrosis through research funding and disease advocacy

Our VALUES

Collaboration
Ethical Decision-Making
Science-Driven

Rosenau Family
Research Foundation

2025 Team

Staff

Gabriel M. Cohn, MD, MBA
Executive Director &
Medical Director

Heather Techmeier
Finance Director

Contractors

Renea Muellerleile
Fractional Operations Director,
Hood & Associates

Rachel Jackson
Fractional Marketing Director

Heidi Carrozzella
Fractional Marketing Leader,
Zella Marketing

Board

Paul Rosenau
President & Co-Founder

Marci Sontag
Chair

Amanda Post
Secretary

Gillian Hauboldt
Treasurer

Phil Christianson
Director

Heather Techmeier
Director

Gabriel M. Cohn
Director

Li Ou
Director

Karlita Blackwell
Director

Teri Cannon
Director

Anne Rugari
Director

Nick Pilger
Director

Zachery Leeker
Director

Gregory Potter
Director

Fresh Voices, Shared Vision: Meet Our New Board Directors

BOARD GOODBYES

The Rosenau Family Research Foundation (RFRF) Board of Directors is tasked with supporting the activities of the Foundation by overseeing the growth of the Foundation's scientific grant portfolio and staff development.

The board continued to grow in 2025, adding five new directors to its ranks while also saying good-bye to two of its valued members, Phil Christianson and Karlita Blackwell.

Phil Christianson
Director

Karlita Blackwell
Director

Phil served as a director in his four years with RFRF. His substantial knowledge of the health care industry and experience with the improvement of mid-sized companies and organizations contributed greatly to RFRF's continuing success in growing our grant program into what it is today.

Karlita served for three years as a director on the RFRF board. As the mother of a child with a rare disease, her extensive experience with patient advocacy was invaluable in guiding the work of the board and the direction of the Foundation, especially in the area of Newborn Screening. We are grateful for her time and knowledge on both a personal and professional level.

We wish them both the utmost best as they continue on their career journeys!

WELCOMING NEW MEMBERS

Joining the board in Phil and Karlita's stead are five new directors who bring a wide range of expertise and knowledge to an already stellar group. Their perspectives will be crucial in guiding RFRF in its efforts to improve the lives of patients impacted by Krabbe disease and Cystic Fibrosis through research funding and disease advocacy.

Read on to learn more about each of our new board directors.

NEW BOARD DIRECTORS IN 2025

Teri Cannon
Director

Teri Cannon joined the RFRF Board in 2025, bringing more than 34 years of experience in nonprofit fundraising and patient advocacy. Her extensive background includes executive leadership roles with the National Multiple Sclerosis Society, Leukemia & Lymphoma Society, Children's Minnesota Hospital, and Greater Minneapolis Crisis Nursery. Teri is currently serving as the executive director of A Breath of Hope Lung Foundation. In addition to her professional leadership, she has also served on boards supporting community health and charitable organizations.

Nick Pilger
Director

Nick Pilger currently works as a development director of The Foundation at Children's Wisconsin, where he works closely with some of southeast Wisconsin's most generous healthcare philanthropists.

Nick holds a B.S. in Nonprofit Management and Human Resources from the University of Minnesota – Carlson School of Management, where he formerly served on two alumni boards for many years. Professionally, Nick has been working in the nonprofit sector for over 15 years, serving in numerous fundraising leadership roles for healthcare and higher education organizations, both in Minnesota and Wisconsin.

Anne Rugari
Director

Anne Rugari was first introduced to Krabbe disease in 1986 when her infant son was diagnosed after months of neurologic decline. In 1999, she had a daughter who was diagnosed at two weeks of age, becoming the fourth newborn in the world to receive a transplant for Krabbe. Anne's diverse experiences, along with her passion to advocate for better treatments, have given her opportunities to work with many Krabbe disease stakeholders to assist in moving the science forward.

Anne's journey with Krabbe disease led her to establish Partners For Krabbe Research (2012), and she is the co-founder of KrabbeConnect (2017). She is a consultant for the Brain and Tissue Bank (2015), working with families wishing to donate their loved one's brain and tissues for research.

She has been involved with supporting families and caregivers as a patient engagement manager; has written two children's books about rare disease; was chosen Rare Disease Difference Maker by Engage Health, and is a consultant and dvocate in the rare disease community.

Zachery Leeker
Director

Zac's first experience with Krabbe disease was in 2000 when his brother, Trevor, was diagnosed at 10 months of age with early infantile Krabbe disease. Though he was only six years old when his brother passed, an advocacy journey was sparked. For more than 25 years, the family dedicated their vacations and free time to advancing awareness and advocacy for Krabbe disease. Annual family trips often centered around attending medical symposiums to learn more about the disease, while summers were devoted to hosting "Trevor's Tournament," a charity softball event that raised scholarship funds for students pursuing careers in the medical field. Much of their remaining time was spent advocating alongside state representatives to expand newborn screening initiatives.

Zac's educational background includes a Bachelor's degree in Kinesiology and a Master's Degree in Healthcare Administration. After years of service on the Kansas Newborn Screening Advisory Council and a short stint in local public health, Zac took over as the program manager for newborn screening in his home state of Kansas. Three years later, Zac was presented with an offer to transition as program manager for Newborn Screening for the state of Iowa and help lead a quad-state collective of screening programs, including North Dakota, South Dakota, and Alaska.

Gregory Potter
Director

Greg is currently the associate director of molecular biology at Precigen Therapeutics, where he leads research programs developing next-generation gene and cell therapies. Previously, he was a scientist at Denali Therapeutics, where he helped advance a small-molecule therapeutic program from the lab bench into clinical trials. Over the past 25 years, he has built a career that bridges academia and industry, with expertise spanning neuroscience, immunotherapy, biologics, and drug development. Greg received his PhD in Neuroscience from Johns Hopkins University School of Medicine.

During his postdoctoral work at the University of California, San Francisco, Greg discovered the underlying mutation in a novel Krabbe disease mouse model. He and his wife later established a small research group at Oregon Health & Science University in Portland, Oregon, where they continued to investigate the molecular mechanisms and disease processes of Krabbe disease using this model. As part of this work, Greg's group received a research grant from The Legacy of Angels Foundation (now RFRF), funding that was instrumental in supporting their lab. He remains grateful for that support and is honored to return to the Foundation as a board member. Greg brings a combined scientific and industry perspective to support strategic planning and to help accelerate therapies for rare diseases.

2025 Investment Committee

The Investment Committee (IC) for Rosenau Family Research Foundation (RFRF) was established in September 2023 to provide recommendations to the board of directors (BOD) for fiduciary oversight and governance of the Foundation's investment assets. The committee is not responsible for directly managing investments; rather, it ensures that a sound and secure framework is in place for prudent investment management. The Foundation's assets are professionally managed by an independent investment manager, Foster Group, under the supervision of the IC.

The Investment Committee's primary responsibilities include:

- Reporting investment performance to the BOD quarterly
- Recommending investment policies for consideration and approval by the board
- Providing recommendations to the board regarding the selection, retention, and evaluation of the external investment manager
- Monitoring financial markets and economic conditions
- Staying informed of regulatory developments affecting investment assets
- Communicating relevant investment updates and insights to the BOD
- Performing any additional duties assigned by the board from time to time

The Investment Committee is composed of the following designated advisors who collectively provide oversight, expertise, and stewardship in support of the Foundation's long-term financial objectives:

Gillian Hauboldt

Gillian Hauboldt earned her Bachelor of Science in Accounting from the University of Wisconsin -Milwaukee. She successfully passed the Uniform CPA Exam and brings more than 25 years of experience in public accounting, with a strong focus on serving nonprofit organizations. Her expertise includes tax and accounting, and her work with nonprofits has also provided her with experience in grant writing.

Gillian is actively involved in the community, serving on several nonprofit boards. She has served as board treasurer since September 2022 and Investment Committee Chair since its inception in September 2023 for Rosenau Family Research Foundation, where she contributes her financial expertise and strategic insight to support the organization's mission.

Mathew Kuhnau

Mathew Kuhnau has served in an advisory role with Edward Jones since 2003 in Greendale, Wisconsin. He earned a Bachelor of Science in Sociology from the University of Wisconsin – La Crosse in 1997 and an MBA from the University of Phoenix in 2003. He also holds the Accredited Asset Management Specialist (AAMS) designation, awarded by the College for Financial Planning, which reflects advanced training in investment planning, portfolio strategy, ethics, and ongoing professional development.

Matt joined the Investment Committee in September 2023 upon its inception and is committed to thoughtful oversight, prudent decision-making, and ensuring investment strategies remain aligned with the organization's mission and long-term sustainability.

Philip Reiter

Philip Reiter has served as a wealth management advisor with Northwestern Mutual in Rochester, MN, for 21 years. He began his career after earning a degree in Agricultural Business from the University of Minnesota. Phil serves on many boards. In 2025, he joined Rosenau Family Research Foundation's Investment Committee, further expanding his involvement in philanthropy and investment stewardship.

Raised on a family farm in rural Elgin, Minnesota, Phil developed a strong work ethic and a deep appreciation for community and family, values that continue to guide both his personal and professional life.

Heather Techmeier

Heather Techmeier has served as the finance director for Rosenau Family Research Foundation since 2018 and has been a dedicated member of the organization's board of directors and Investment Committee since its founding. With more than 20 years of corporate finance experience and eight years in nonprofit financial management, she brings a strong foundation of expertise in capital banking, cash management, financial reporting, data analysis, investments, and budgeting. Her leadership plays a vital role in maintaining and safeguarding the financial stability and long-term sustainability of the Foundation.

Heather's commitment to RFRF is deeply personal. Her life has been directly impacted by genetic testing, which revealed she is a carrier of two autosomal recessive conditions—Krabbe disease and Cystic Fibrosis. She also experienced the profound loss of her niece, Makayla Pike, who passed away from Krabbe disease in May 2003. These experiences have strengthened her dedication to advancing research and disease advocacy for families affected by rare genetic conditions.

Beyond her work with RFRF, Heather is actively involved in several nonprofit organizations, including the Center for Public Health Innovation (CPHI), the Muskego Wrestling Club, and the Christmas Clearing Council, among others. Her community engagement reflects her ongoing commitment to service and impact.

Bradley Vick

Bradley Vick brings more than 35 years of experience in the financial services industry, establishing a long-standing reputation for knowledge, professionalism, and client-focused wealth management. He holds the Certified Financial Planner (CFP) certification, the Chartered Financial Analyst (CFA) designation, and the Wealth Management Certified Professional (WMCP) designation, reflecting a deep commitment to advanced financial planning and investment experience. He is currently a private wealth advisor with LPL Financial, where his approach emphasizes disciplined planning, informed investment management, and a strong dedication to client relationships.

In addition to being on RFRF's Investment Committee since 2025, Brad is actively engaged in a range of charitable organizations, including Semper Fi & America's Fund, Zero Prostate, Unbound Global, and Blood Cancer United. His involvement reflects a commitment to giving back and supporting causes that make a meaningful difference in the lives of others.

2025 Market Review and Portfolio Update

Heather Techmeier
Finance Director

Throughout the year, equity markets experienced periods of turbulence, particularly in early spring when concerns around trade policy and tariffs created short-term uncertainty. However, these disruptions proved temporary. By year-end, U.S. equities posted gains exceeding 17 percent, demonstrating steady recovery and overall strength.

Even more notable was the performance of international and emerging markets. International equities rose by more than 32 percent, while emerging markets delivered gains exceeding 33 percent. This marked the first time since 2022 that international equities outperformed their U.S. counterpart, highlighting the cyclical nature of global market leadership. Fixed income also contributed positively, with U.S. bonds returning over 7 percent, supported by modest declines in interest rates and consistent income generation (see Graph A).

Graph A

Year to Date Asset Class Returns

(as of 12/31/25)

"PAST PERFORMANCE IS NO GUARANTEE OF FUTURE RESULTS. Please see specific disclaimer that follows. Results do not reflect the impact of the deduction of management fees, nor do they include the impact of custodial or transaction fees."

Source: Bloomberg. Indexes used are Global Stocks: MSCI ACWI IMI, US Large: Russell 1000 TR, US Large Growth: Russell 1000 Growth, US Large Value: Russell 1000 Value TR, US Small: Russell 2000 TR, US Small: Russell 2000 Growth TR US Small Value: Russell 2000 Value, Global REIT: S&P Global REIT, Intl Large: MSCI World Ex USA Large NR, Intl Large Value: MSCI World Ex USA Large Value, Intl Large Growth: MSCI World Ex USA Large Growth, Intl Small: MSCI World Ex USA Small NR, Intl Small Value: MSCI World Ex USA Small Value, Emerging Markets: MSCI EM NR & FTSE EM for 20 Yr, US Agg: BBgBarc US Agg Bond TR, US Short-Term: BBgBarc US Govt/Credit 1-3 Yr TR.

One of the defining features of 2025 was the broad-based nature of returns. Virtually all major asset classes finished the year in positive territory. Such synchronized performance across asset classes is relatively uncommon and underscores the benefit of maintaining a diversified investment approach.

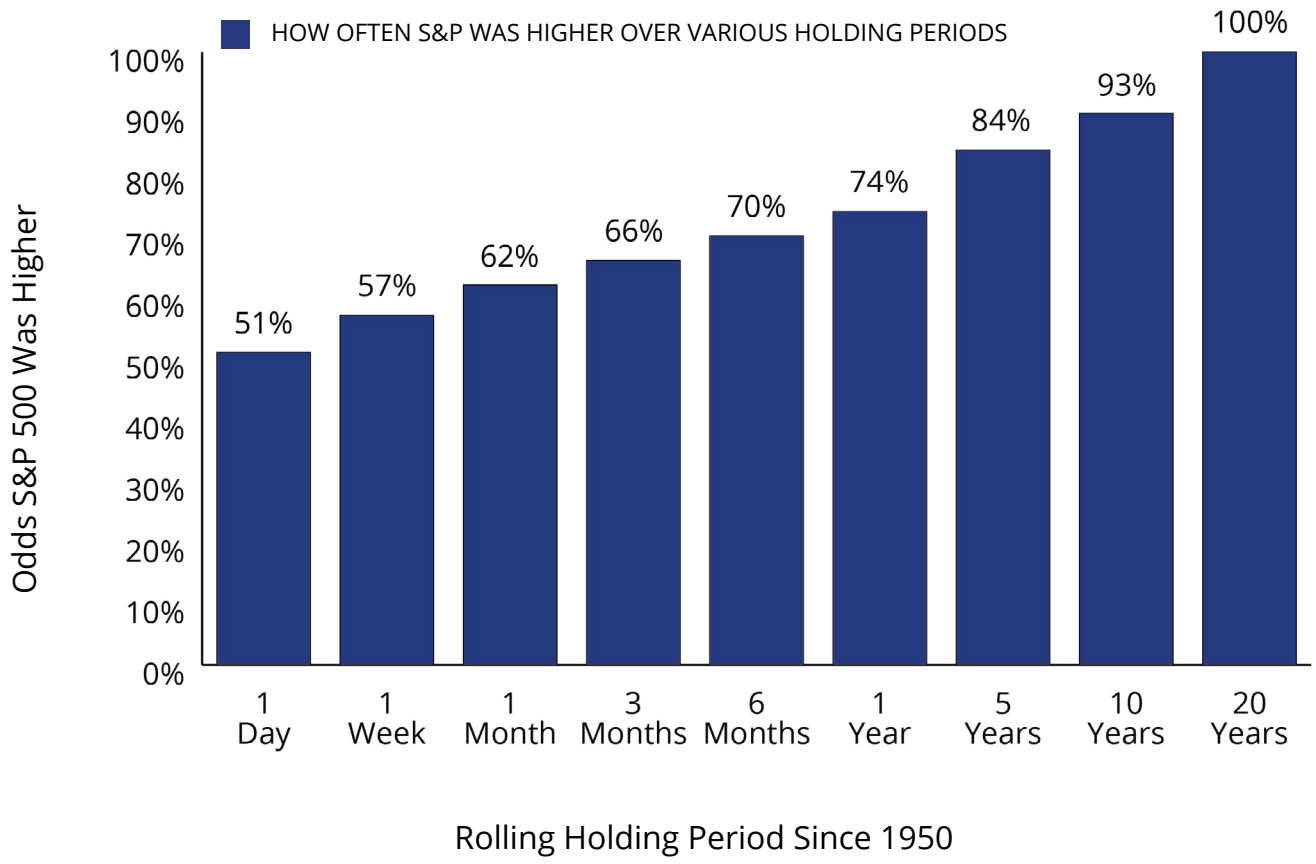
At the same time, market concentration remained an important theme. A small group of large-cap technology companies, often referred to as the "Magnificent 7", continued to exert significant influence on overall market performance. Representing roughly 35 percent of the U.S. equity index and about 40 percent of the top ten companies by market capitalization, these firms drove a substantial portion of both gains and volatility. Notably, this group experienced a significant draw down during the year before ultimately recovering to end in positive territory. This dynamic highlights the importance of diversification, as leadership within markets can shift over time and concentration can introduce heightened risk.

Looking ahead at the wide range of possible outcomes for 2026, the guiding investment philosophy remains unchanged: maintain a disciplined, long-term approach grounded in diversification rather than attempting to time markets or select individual securities. Historical evidence continues to support the principle that time in the market is a more reliable driver of success than short-term positioning (see Graph B).

Graph B

Time in the Market > Timing the Market

Historically, Odds of Positive Returns Increase with Holding Period



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Source: Bloomberg. Indexes used are Global Stocks: MSCI ACWI IMI, US Large: Russell 1000 TR, US Large Growth: Russell 1000 Growth, US Large Value: Russell 1000 Value TR, US Small: Russell 2000 TR, US Small: Russell 2000 Growth TR US Small Value: Russell 2000 Value, Global REIT: S&P Global REIT, Intl Large: MSCI World Ex USA Large NR, Intl Large Value: MSCI World Ex USA Large Value, Intl Large Growth: MSCI World Ex USA Large Growth, Intl Small: MSCI World Ex USA Small NR, Intl Small Value: MSCI World Ex USA Small Value, Emerging Markets: MSCI EM NR & FTSE EM for 20 Yr, US Agg: BBgBarc US Agg Bond TR, US Short-Term: BBgBarc US Govt/Credit 1-3 Yr TR.

2025 Market Review and Portfolio Update

From a portfolio perspective, Rosenau Family Research Foundation (RFRF) remains well-aligned with its long-term objectives. The primary investment goal, to achieve returns exceeding inflation over time, continues to guide strategy and decision-making. As of December 31, 2025, the portfolio was valued at approximately $40.3 million, while also supporting the organization's mission through awarding more than $1.7 million in new and recurring grants to researchers.

Rosenau Family Research Foundation Balance Sheet
As of December 31, 2025

ASSETS

	TOTAL
Current Assets	
Checking Account	$130,945.23
Investment Account	$40,313.746.38
Other Current Assets	$15,190.70
Other Assets	
Life Insurance	$1,868,153.52
TOTAL ASSETS	**$42,328,035.83**

LIABILITIES AND EQUITY

Liabilities	
Current Liabilities	$1,570.22
Total for Liabilities	**$1,570.22**
Equity	
Retained Earnings	$38,085,379.13
Net Income	$4,241,086.48
Total Equity	**$42,326,465.61**
TOTAL LIABILITIES AND EQUITY	**$42,328,035.83**

Asset allocation remains consistent with RFRF's investment policy targets, with approximately 60 percent allocated to growth-oriented investments and 40 percent to preservation assets. This balance has been maintained steadily over time and reflects a commitment to both long-term capital appreciation and short-term stability.

Performance in 2025 was strong, with the portfolio delivering a net return of 16.76 percent after all fees and expenses. Key contributors to performance included U.S. large value equities, momentum-driven strategies, and broad international exposure.

In summary, 2025 was an exceptionally strong year across global markets, reinforcing the values of diversification and long-term investing. While the outlook for 2026 includes a range of possible scenarios, the portfolio remains well-positioned to navigate uncertainty. Future discussions will continue to evaluate asset allocation in light of evolving spending needs and long-term objectives, ensuring the investment strategy remains aligned with the Foundation's mission.

Grants & Research

2025 Grant Recipients

Rosenau Family Research Foundation considers grant funding for basic science research, translational research, and clinical research that can lead to or enhance treatments and cures for Krabbe disease and Cystic Fibrosis. Programs that promote, through education and awareness, the expansion of Newborn Screening, Krabbe disease, and Cystic Fibrosis are similarly considered.

Our 2025 grant recipients showcase the broad spectrum of work being done to push the needle forward towards finding effective treatments and improving the lives of patients living with these diseases.

CLICK ON EACH GRANT RECIPIENT TO LEARN MORE

"ENGINEERING ADENO-ASSOCIATED VIRAL VECTORS FOR BROAD CENTRAL AND PERIPHERAL NERVOUS SYSTEM TARGETING"
Changfan Lin (Caltech) NEW INVESTIGATOR

"MOLECULAR CHARACTERIZATION AND PATHOGENICITY ASSESSMENT OF COMMON VARIANTS OF UNCERTAIN SIGNIFICANCE IN KRABBE DISEASE"
Chris Lee, PhD (Biomedical Research Institute of New Jersey)

"ENRICHING CF NEWBORN SCREENING OUTCOMES THROUGH A PRECISION PERSONALIZED MEDICINE STRATEGY"
Hara Levy, MD, MMSC (University of Wisconsin, Madison)

"GENOKRABBE: A COMPREHENSIVE GENOTYPE-PHENOTYPE DATABASE FOR KRABBE DISEASE"
Robert Thompson Stone, MD (University of Rochester)

"AUTOLOGOUS TRANSPLANTATION OF GENOME-EDITED HEMATOPOIETIC STEM AND PROGENITOR CELLS FOR KRABBE DISEASE"
Natalia Gomez-Ospina, MD, PhD (Stanford University)

"INVESTIGATIING THE ROLE OF A NOVEL TMEM16A ISOFORM IN CYSTIC FIBROSIS"
Thomas Brett, PhD (Washington University in Saint Louis)

"LEVERAGING MICROGLIA REPLACEMENT TOOLS TO UNDERSTAND AND IMPROVE CROSS-CORRECTION IN KRABBE DISEASE"
Venkata Sai Chaluvadi (University of Pennsylvania)

Research Spotlight: Krabbe Disease

In 2023, Rosenau Family Research Foundation awarded a $246,690 grant to KrabbeConnect for its proposed study, "**Parent-Report Disease Burden in Infantile and Late Infantile Krabbe Disease; A Comparison of Quality-of-Life Outcomes in Transplant and Non-or-Late Transplant Patients"**.

Dr. Nicholas Bascou, from Johns Hopkins University, and Stacy Pike-Langenfeld, co-founder and president of KrabbeConnect, served as the study's co-principal investigators. The study was completed in 2024, with results published in 2025.

About the Study

This study addresses the absence of investigations on the impact of hematopoietic stem cell transplantation (HSCT) on quality of life, in particular for caregivers of patients with Krabbe disease (KD).

Investigators developed a 90-minute caregiver interview to gather qualitative and quantitative data (including the validated Leukodystrophy Quality-of-Life Assessment – LQLA). This interview was designed to explore the following: 1) disease burden on the patient; 2) physical burden on the caregiver; and 3) emotional/social burden on the caregiver. Comparisons were made between children not transplanted/transplanted late and children transplanted early. Infantile KD (IKD) and late infantile KD (LIKD) were analyzed independently.

Study partners Dr. Nicholas Bascou, Stacy Pike-Langenfeld, and Patti Engel, CEO of Engage Health, at WORLDSymposium 2025

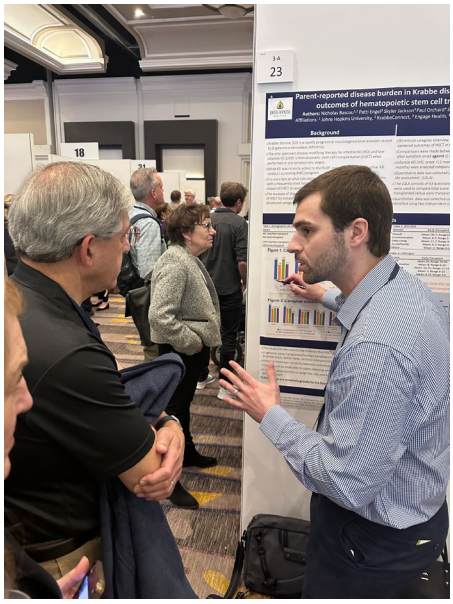
Findings

Analysis of the LQLA revealed that early transplantation led to a relative reduction in disease burden for both IKD and LIKD groups. For the IKD group, there was a tendency towards decreased physical burden on caregivers of children transplanted early. Although all groups experienced significant social/emotional burdens, caregivers of IKD transplanted early benefited from improved sleep, mental health, and familial/spousal relationships compared to IKD non-transplanted/transplanted late.

Of special interest in the study findings were the qualitative results from the interviews. 100 points were given for each of the three categories listed above, and the interviewer could distribute those points however they saw fit, giving quantifiable measurements to the more nuanced and open-ended responses.

One finding was that the physical burden was less on the caregiver in the early transplant group, which was expected, but caregiver responses also showed that as transplant children grew older, the physical burden became more significant on the caregiver. This was because transplantation helps cognitive and fine motor skills, but not gross motor skills, so as children grew up, caregivers had to do more lifting.

Findings, cont.



Dr. Nicholas Bascou presenting a poster at WORLDSymposium 2025

Another finding from the qualitative interview questions was that the caregivers of non-transplant and transplant late patients were more focused on basic survival and daily needs. They also tended to have higher levels of psychiatric stress (depression, anxiety, hopelessness, and guilt).

In the early transplant group, caregivers were found to be able to transcend focusing on just basic needs. They were able to move beyond survival mode and strive for more social and emotional well-being, to enhance their quality of life. There were still hardships and struggles, but they had lower levels of psychiatric stress.

These findings highlighted the importance of qualitative data in caregiver studies since they weren't reflected in the quantitative responses, but emerged when caregivers were given the opportunity to speak about their experiences in more nuanced ways.

Moving Forward

This study provides collective wisdom from parents and caregivers who have gone through a similar experience to those just learning about their child's KD diagnosis.

Its findings show convincing evidence that HSCT improves quality of life and reduces caregiver burden in IKD. This data will be critical in the decision-making process for states not currently screening for KD but debating the addition of KD to their NBS panels.

The findings from this study will also help families to weigh the risks and benefits of HSCT more confidently when contemplating the life-altering decision of whether to proceed with transplantation.

Additionally, the investigators are interested in publishing a paper focusing on the irritability cry of babies with KD, to give caregivers and doctors specific information on what to be listening for in order to differentiate it from other conditions and hopefully lead to an even earlier diagnosis. They are also looking into developing a scale or a rating system for caregiver burden, similar to a mental health survey that patients often fill out before doctor visits, which would help track how a caregiver is doing over time.

Click the button below for more information and study results.

Read the Full Manuscript

Research Spotlight: Cystic Fibrosis

Thomas Brett, PhD
Washington University in St. Louis

About the Study

In 2025, Rosenau Family Research Foundation awarded a $375,000 grant to Dr. Thomas Brett, and collaborator Dr. Jen Alexander-Brett, for their project titled **"Investigating the role of a novel TMEM16A isoform in cystic fibrosis"**. The main focus of this project is studying how a chloride channel in the airway, called TMEM16A, can be targeted in the setting of Cystic Fibrosis (CF) to restore beneficial mucus function in CF patients. They discovered a specific potentiator, termed CLCA1 VWA, that can selectively potentiate TMEM16A, which improves the impaired mucociliary transport seen in CF patient samples. Targeting an alternative channel (separate from the cystic fibrosis transmembrane conductance regulator, or CFTR) could represent a universal treatment or supplement for CF patients that would not be dependent on their CFTR mutation (of which there are thousands). Their work has also identified a novel form of TMEM16A, called $TMEM16A_{acd}$, that they found is expressed in CF patient airways and may be a more active form of the channel.

2025 Progress

So far their work shows that $TMEM16_{acd}$ is more highly expressed in Cystic Fibrosis patients than it is normally in healthy populations. This means that this form of the channel may be a strong target to improve mucus properties in CF patients. The team has also embarked on ground-breaking genetics studies, called spatial transcriptomics, which can identify where in CF lung tissues TMEM16A is expressed, and how its patterns might change with disease settings.

> ***"The generous funding from Rosenau Family Research Foundation has fueled our foray into an exciting new frontier in studying CF; using state-of-the-art spatial transcriptomics to study an increasing important channel in the airway, TMEM16A. The work could lead to therapeutics that improve mucus function in CF and other diseases hallmarked by mucus obstruction, like COPD."***
>
> - Tom Brett

Moving Forward

The team will characterize $TMEM16_{acd}$ to understand how it can be targeted in CF. Spatial transcriptomics studies will be used not only to understand where TMEM16A is expressed, but also to understand how its expression location can change with disease, and also to identify other partner proteins which might assist in modulating its activity. In the end, they hope to demonstrate that TMEM16A is a beneficial chloride channel to target in CF, as well as other mucus-obstructive diseases like COPD and severe asthma.

Learn More About This Study

2025 Reports & Publications

MARCH 2025

An article was published by **Philip Farrell, MD, PhD,** and team in *Pediatric Pulmonology*, based on the whole genome sequencing studies completed with funding from The Legacy of Angels Foundation, now Rosenau Family Research Foundation (RFRF), on the first cohort of children with Cystic Fibrosis who were diagnosed through screening.

READ THE MANUSCRIPT

APRIL 2025

A landmark article was published by **Philip Farrell, MD, PhD**, and team in the *International Journal of Neonatal Screening*, outlining the new Cystic Fibrosis Foundation's recommended best practice guidelines for Cystic Fibrosis newborn screening. It is the product of three years of intensive work, and can be partially attributed to a grant awarded to **Mei Baker** by The Legacy of Angels Foundation, now RFRF, titled "**A Prospective Study of Newborn Screening for Cystic Fibrosis Using a Novel IRT/Next Generation Sequencing"**. Of note, co-authors include **Susanna McColley, MD**, and RFRF's Board of Directors Chair, **Marci Sontag, PhD**.

READ THE MANUSCRIPT

MAY 2025

Susanna McColley, MD, and her team at Lurie Children's Hospital published a manuscript in *Pediatric Pulmonology* with findings from research supported by an RFRF grant titled "**Caregiver and Clinician AwaREness for early Cystic Fibrosis diagnosis (C-CARE CF)"**.

READ THE MANUSCRIPT

JUNE 2025

RFRF Executive Director **Gabriel M. Cohn, MD, MBA**, was featured on the Living With Cystic Fibrosis podcast, hosted by Laura Bonnell, President of The Bonnell Foundation.

LISTEN TO THE PODCAST

JULY 2025

Diego Zelada, **Natalia Saldivia** and **Maria Irene Givogri, PhD**, with others, published a manuscript in the *International Journal of Nanomedicine* with findings related to an RFRF grant awarded in 2020 titled "**Vesicular Delivery of GALC for ERT of Krabbe's Disease"**.

READ THE MANUSCRIPT

2025 Lab Tour:
Biomedical Research Institute of New Jersey

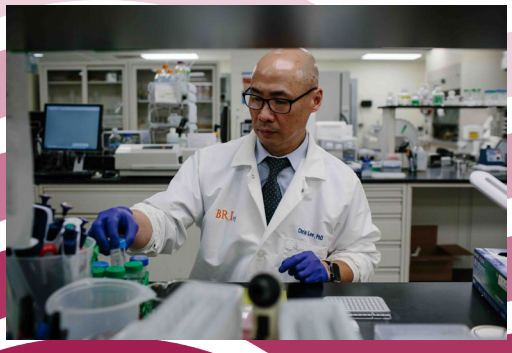
In August 2025, Rosenau Family Research Foundation (RFRF) Fractional Marketing Director Rachel Jackson, and a two-person film crew from RFRF's digital marketing partner, ArcStone, took a trip to Cedar Knolls, New Jersey, to visit grant recipient Dr. Chris Lee and his team at the Biomedical Research Institute of New Jersey (BRInj).

Dr. Lee gave the team a tour of his lab and sat down to talk about discovering a love of science when he was a kid, his journey to becoming a scientist himself, and his work in Krabbe disease research utilizing RFRF funding.

His current grant, titled **"Molecular characterization and pathogenicity assessment of common variants of uncertain significance in Krabbe Disease"**, has two goals. The first is to establish a human cell model to understand how DNA mutations cause Krabbe disease, and to classify those mutations to identify which ones are more likely to cause higher severity. The second is to use this cell model as a system to test potential drugs that could be used as treatments for Krabbe disease. Dr. Lee shared some findings from the research that he and his team are very excited about, which you can learn more about by watching the video above.

Dr. Lee and his colleagues at the lab, Dr. Christopher B. Eckman and Dr. Elizabeth E. Eckman, also spoke to the importance of RFRF grant funding in ensuring their project moves forward. These lab tours give RFRF and its network the opportunity to see the Foundation's grant dollars at work, and to share the stories of grant recipients with everyone invested in RFRF's mission of improving the lives of patients impacted by Krabbe disease and Cystic Fibrosis through research funding and disease advocacy.

The team also had the chance to talk with RFRF's first Student Internship Program participant, Paridhi Tyagi! You can learn more about Paridhi and the Student Internship Program on pages 30-31.



Partnerships, Programs & Awards

KTRN
Krabbe Translational Research Network
2025 MEETING

PRESENTED BY
Rosenau Family
Research Foundation

Gabriel M. Cohn, MD, MBA
Executive Director

Krabbe disease remains a devastating leukodystrophy with urgent unmet needs across the entire journey: early detection, confident diagnosis, predicting disease course, selecting the right intervention at the right time, and measuring whether a therapy is truly effective.

In 2025, the Krabbe Translational Research Network (KTRN) convened researchers, clinicians, and community stakeholders around a shared focus: "2030"—a future-facing objective to accelerate progress toward better diagnosis, care, and treatments for Krabbe disease.

"KTRN 2030" consisted of scientific sessions, comprised of scientific platform presentations and several expert panel discussions. The meeting's core goal was to identify the challenges, gaps, and needs of people impacted by Krabbe disease—and the scientific and infrastructure priorities that must be addressed to close those gaps by 2030.

Across scientific sessions and panel discussions, the meeting balanced practical near-term needs (like harmonizing newborn screening follow-up and standardizing care pathways) with the deep biology required to unlock next-generation therapeutics. Just as importantly, the meeting emphasized collaboration: building shared resources, aligning on common standards, and strengthening the connections that turn discoveries into real-world impact.

SCIENTIFIC SESSIONS

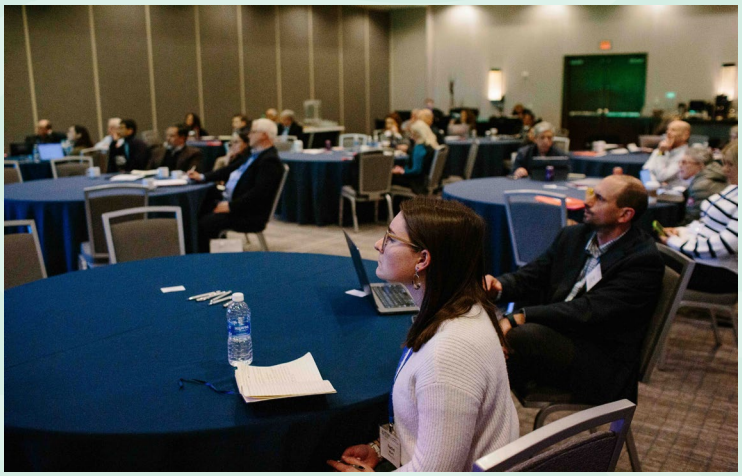
These sessions highlighted progress in disease models, biomarkers, and clinical insights—each essential to building better therapies and better care.

New experimental systems and disease models were presented to better characterize prevalent-related mutations and to study disease mechanisms more precisely. Of note were new mouse models described as developing disease earlier than the commonly used Twitcher (TWI) model, offering new windows into neurological features including the hippocampus (important for learning and memory). Earlier-onset models can shorten testing timelines for candidate therapeutics and may capture features not well represented in existing systems. Alternatively, additional model work aimed at studying later-onset forms of Krabbe disease—critical because the disease spectrum is broader than the classic infantile presentation, was discussed.

A compelling line of discussion focused on **cerebrospinal fluid (CSF) dynamics and biology** in Krabbe disease. Findings described CSF enriched with immunoglobulin-related peptides and other proteins consistent with an antibody response, suggesting an overactive immune component—specifically elevated B-cell–associated activity in the dura mater. For experts, this points to immune pathways that may meaningfully intersect with neuroinflammation and disease progression and may offer new biomarkers or potential therapeutic targets.

The meeting also highlighted **metabolomics** approaches—methods that measure small-molecule "signatures" in blood plasma—which may enhance diagnostic capabilities and help clarify ambiguous cases. This is particularly relevant for the real-world problem of uncertain results in screening and follow-up, where more precise biomarker tools are urgently needed.

Clinical presentations discussed evidence that **hematopoietic stem cell transplantation (HSCT)** can improve quality of life and reduce caregiver burden in infantile Krabbe disease (IKD). Another practical clinical observation shared was that infants with IKD may have a unique and identifiable cry potentially supporting earlier recognition and diagnosis. These insights, while not substitutes for definitive testing, may support more timely diagnosis and intervention.



PANEL DISCUSSIONS

A major theme across the expert panels was that scientific progress will move faster—and therapies will reach people sooner—when the community builds shared, open resources and aligns on standards.

Basic Science Research: For experts, the meeting highlighted several research priorities that map directly to future therapeutic strategies and clinical translation. These included advancing our understanding of pathogenic mechanisms, including the role of immunity and how disease unfolds during in utero brain development, and the role of psychosine in disease development along with the potential consequences of over-treatment, further underscoring the need for precision in both biomarkers and interventions.

Translational Science Research: Developing and validating large animal models to complement cell and mouse systems were cited as approaches to improve translational confidence. Building cell models and functional assays that can accurately predict the pathogenicity of GALC variants may accelerate variant classification, support clinical trial eligibility decisions and support therapeutic development.

Clinical Research-Centralized Natural History and Biorepository: Some of the strongest consensus was a need for a centralized natural history effort to advance the understanding of Krabbe subtypes and their trajectories. For families, "natural history" means capturing what the disease looks like over time—symptoms, milestones, tests, imaging, outcomes, and the impact of care. For experts, a robust dataset enables better subtype stratification, more accurate prognosis, and stronger clinical trial design—especially for rare diseases where every data point matters. Importantly, such a database can support evaluation of the safety and efficacy of interventions and help the field agree on meaningful endpoints. Complementing the database discussion was a call for a centralized biorepository—a coordinated collection of biospecimens linked to high-quality clinical data. The group emphasized that this resource could enable better diagnostic, prognostic, predictive, and surrogate biomarkers, refine Newborn Screening approaches, and support variant interpretation—including variants of uncertain significance (VUS). For drug development, a biorepository could accelerate biomarker validation, strengthen regulatory conversations, and support trial readiness and even accelerate approval pathways when paired with credible surrogate endpoints.

Panels stressed the importance of combining natural history studies, family histories, biomarker profiles, and genetic analysis to better define Krabbe subtypes and disease course. A recurring challenge is the diagnostic and prognostic uncertainty surrounding "grey zone" biomarker results and VUS. The meeting reinforced that solving this problem is not a single-test issue—it requires integrated datasets, functional assays, and shared interpretation frameworks.

Clinical Research - Therapeutics: The sharing of updates and lessons learned across gene therapy clinical trials were considered critical as was the exploration of additional modalities (including immune-focused approaches and small molecules) either as standalone therapies or as bridges to transplantation.

Clinical Considerations:

Improvements in Newborn Screening and Standardized Care: Panelists emphasized the need to add Krabbe disease onto Newborn Screening (NBS) panels across all 50 states, alongside improving the systems that connect state labs, healthcare providers, and families. Beyond the addition of screening, the meeting stressed the importance of delivering NBS information accurately, sensitively, and through appropriate medical experts so that families receive timely, expert-guided care regardless of location. Mechanisms must also be in place to ensure that the information is acted upon in a timely manner and that follow-up is tightly coordinated to ensure that additional follow-up testing, referrals, and expert care can happen quickly.

The need for standardized care of patients identified through NBS was stressed and includes what families are told, how confirmatory testing proceeds, and how care is coordinated. Proposed solutions included practical "toolkits," improved provider education (especially for pediatricians and clinicians delivering NBS results), and stronger networks that connect local providers with regional Krabbe experts.

Consensus on Transplantation and Outcomes Tracking: Panelists called for greater consistency around transplant-related decisions, including refining conditioning regimens (with interest in reduced-intensity approaches) and formalizing how outcomes are collected over time. Systematic outcomes tracking is essential both for families making decisions today and for a field trying to compare interventions rigorously.

Strengthening the pipeline of investigators by attracting younger scientists and clinicians into the field and encouraging cross-talk between laboratory and clinical communities.

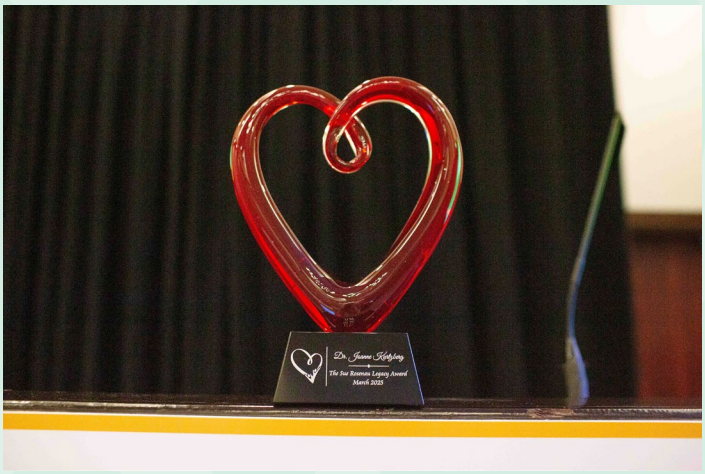
KTRN
Krabbe Translational Research Network
2025 MEETING

PRESENTED BY
Rosenau Family
Research Foundation

The 2025 KTRN meeting made clear that progress in Krabbe disease is not constrained by a single bottleneck. Instead, success depends on synchronizing multiple efforts: stronger models, clearer biomarkers, shared datasets and samples, coordinated newborn screening follow-up, standardized care, and a broad therapeutic pipeline. The meeting's discussions pointed toward a future where families receive faster, expert- guided answers and advice, clinicians have clearer guidance, and researchers can move candidates through the pipeline with more confidence and shared infrastructure. Just as important, the high satisfaction and perceived value of the meeting, as reported by the meeting participants, reflects a community that is aligned, motivated, and ready to build together. The work ahead is ambitious—but the roadmap is sharper than ever, and the network's talent and collaborative energy are the strongest assets we have. During KTRN 2026 we will expand the panel discussions to include all KTRN attendees and work together to invent a future where individuals will no longer have to live life defined by Krabbe disease.

2025 Sue Rosenau Legacy Award

The Sue Rosenau Legacy Award (SRLA) was also handed out at the annual KTRN meeting. The SRLA recognizes an inspirational leader who has collectively created lasting change and measurable differences in one or more areas of RFRF's mission. At the KTRN 2025 meeting, RFRF awarded Joanne Kurtzberg, MD with the 2025 SRLA. Dr. Kurtzberg is an internationally recognized pediatric hematologist–oncologist and transplant physician at Duke University whose career has transformed the outlook for children with Krabbe disease and other inherited metabolic disorders. As founder and director of Duke's Pediatric Blood and Marrow Transplant Program and one of the pioneering leaders in umbilical cord blood transplantation, Dr. Kurtzberg helped establish stem cell transplantation as a life-altering—and often lifesaving—therapy for infants diagnosed early with Krabbe disease. Her decades of clinical innovation, advocacy for newborn screening, and unwavering commitment to translating research into patient care have collectively created lasting and measurable impact. Through her leadership, mentorship, and compassion for families, Dr. Kurtzberg embodies RFRF's mission and is a fitting recipient of the 2025 Sue Rosenau Legacy Award.



Student Internship Program

Rosenau Family Research Foundation (RFRF) is invested in engaging young, talented minds who are interested in rare disease research and advocacy and recruiting them to focus their studies on the fields of Krabbe disease and Cystic Fibrosis. In 2025, the Foundation created the Student Internship Program to provide funding for internship opportunities that are in need of financial support. The Foundation's hope for this program is to assist in administering internships that might not otherwise be able to happen due to financial restrictions.

KEY POINTS ABOUT THE STUDENT INTERNSHIP PROGRAM

- Three awards of $5,000 each are available annually on a first come, first served basis
- Sponsors (faculty, advisors, etc.) can nominate a student (high school through PhD) for a 6-12 week Krabbe disease or Cystic Fibrosis research internship
- Sponsors must be the ones to apply and provide a general outline for the internship
- One award is given per sponsor

If you are interested in discussing an internship with RFRF, Please click the button below, and email racheljackson@rosenaufoundation.org with any questions.

Apply for student internship funding

Student Internship Program

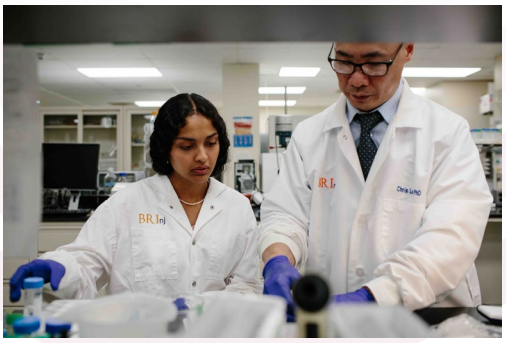
Biomedical Research Institute of New Jersey

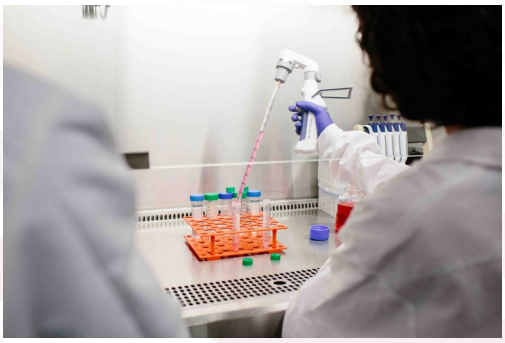
While in New Jersey filming a tour and interviews with Dr. Chris Lee at his Biomedical Research Institute of New Jersey (BRInj) lab in August, the RFRF team had the opportunity to interview the first participant in its Student Membership Program, Paridhi Tyagi.

Paridhi joined Dr. Lee and his team for a two-week internship to assist them with work on their RFRF-funded grant, which you can learn more about on page 21.

An incredibly bright and ambitious seventeen-year-old, Paridhi spoke about what inspired her to pursue rare disease research, and her excitement about being able to move beyond her classroom textbooks and get hands-on, applicable experience in a lab, especially as a high school student who might not be presented with as many internship options as students at a university or other higher educational institution. The team also interviewed Dana Clausen, BRInj's Student Intern Project Leader, who echoed the importance of providing high schoolers with an avenue to follow their curiosity and passion.

Paridhi's internship with BRInj was the perfect showcase of why RFRF developed the Student Internship Program in 2025. The Foundation is looking forward to seeing other internships follow, both at the high school level and beyond. Click on the video above to learn more about Paridhi's experience, and again, please reach out if you have an internship opportunity you would like RFRF to help fund.





2025 Event Sponsorships

In 2025, Rosenau Family Research Foundation sponsored a variety of events. Supporting the rare disease community is a crucial part of the Foundation's work each year as it strives to fulfill its mission of improving the lives of patients impacted by Krabbe disease and Cystic Fibrosis through research funding and disease advocacy.

KrabbeConnect
CONNECTING PATIENTS & RESEARCH

SWING FOR SAYLOR GOLF TOURNAMENT, *June 1st*
FAMILY WARRIOR MEETING, *June 28th*
BLOOM FOR A CURE GALA, *August 23rd*

NORD®
National Organization for Rare Disorders

STUDENTS FOR RARE PROGRAMMING
NORD SUMMIT, *October 19th - 21st*

HUNTER'S HOPE

MEDICAL & FAMILY SYMPOSIUM, *July 14th - 20th*

THE BONNELL FOUNDATION:
LIVING WITH CYSTIC FIBROSIS
LIVE • BREATHE • INSPIRE
Roadmap to CF™ | thebonnellfoundation.org

HARVEST OF HOPE GALA, *September 20th*

CYSTIC FIBROSIS FOUNDATION®

CORKS & KEGS, *November 21st*

Connect With Us!

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