Rosenau Family
Research Foundation

July
2025
Newsletter

In This Edition

Podcast Feature

Earlier this spring, RFRF’s Executive Director, Gabriel Cohn, chatted with Laura Bonnell of The Bonnell Foundation on her podcast, Living With Cystic Fibrosis. The episode was released in June, alongside a lovely write-up from Laura, which you can read below.

Click to listen to the episode:

Personalized Mission: The Rosenau Family Foundation story

JUNE 15TH, 2025 | 34:10 | S3:E126

“‘Oh, the people you’ll meet, and the places you’ll go…’

That classic Dr. Seuss line couldn’t have been more true when I found myself at a rare disease conference, taking with Dr. Gabriel Cohn. A quiet presence with a resume that reads like a roadmap through the last 30 years of rare disease breakthroughs, Dr. Cohn is the kind of person who reminds you just how much heart and science it takes to change lives.

Dr. Gabe Cohn is the Executive Director and Medical Director of the Rosenau Family Research Foundation. He stepped into the role in October, bringing with him over three decades of experience that spans academic medicine and the biotechnology industry. A licensed, board-certified Clinical Geneticist and Obstetrician-Gynecologist, Dr. Cohn has contributed to the development of multiple therapeutics aimed at treating rare genetic disorders—a career built on both clinical precision and compassionate innovation.

Beyond RFRF, he’s also the Chief Medical Officer at iECURE, Inc., a biotech company pioneering gene editing therapies for rare diseases. His prior leadership roles at Homology Medicines, AVROBIO, OvaScience, and Shire reflect a steady focus on advancing gene therapy and editing platforms. Since 2017 alone, he’s played a pivotal role in getting five different cell and gene therapy programs past the critical regulatory gatekeeping stages of IND and CTA submissions.

Dr. Cohn isn’t just a scientist—he’s a builder, a connector, and a relentless advocate for the potential of genetic medicine to rewrite the future for patients with rare diseases.”

Grant Recipient Interview: Rachel Wurth

Rachel Wurth, M.S., (MLS) ASCPCM is a PhD candidate in the Clinical and Translational Science Track at Mayo Clinic Graduate School of Biomedical Sciences. She has designed and implemented an untargeted metabolomics platform that is broadly useful in characterizing the small molecule signature of inborn errors of metabolism for the purpose of biomarker discovery. The project focuses on characterizing the metabolic signature of Krabbe disease, a rare and devastating inherited neurological disease. Through partnership with the clinical laboratory, Rachel aims to translate discovered biomarkers into novel diagnostic assays that can improve the detection and prognostic monitoring of individuals with Krabbe disease.

Rachel was awarded $112,718 Rosenau Family Research Foundation in 2024 for a New Investigator Grant titled “Elucidating the metabolic signature of Krabbe Disease plasma, dried blood spots, and cerebrospinal fluid using untargeted metabolomics and metabolic flux analysis”. We sat down with Rachel to discuss what drew her to the field of rare disease research, the challenges and joys of her work, and more.

Watch the Interview:

Medical Student Internship Program

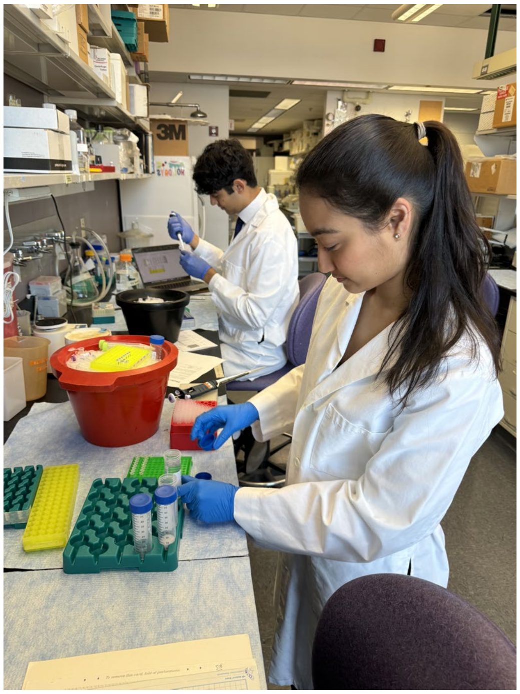
Rosenau Family Research Foundation is invested in engaging with young, talented minds who are interested in rare disease research and advocacy, and recruiting them to focus their studies on the field of Krabbe disease.

About our Medical Student Internship Program:

- Five awards of $5,000 each are available annually
- Sponsors can nominate a medical student (at any level) for a 6-12 week internship
- One award is given per sponsor

If you are a sponsor interested in discussing a student internship with us, click the button below.

Inquire About Student Internship



Grant Research Findings

Parent-reported disease burden in Krabbe disease: Evaluating outcomes of hematopoietic stem cell transplant

Authors: Nicholas Bascou[1,2] Patti Engel[2] Skyler Jackson[3] Paul Orchard[4] Stacy Pike-Langenfeld[2]

Affiliations: [1] Johns Hopkins University, [2] KrabbeConnect, [3] Engage Health, [4] University of Minnesota

Background

- Krabbe disease (KD) is a rapidly progressive neurodegenerative disorder caused by β-galactocerebrosidase deficiency.
- The only approved disease modifying therapy for infantile KD (IKD) and late-infantile KD (LIKD) is hematopoietic stem cell transplantation (HSCT) when performed in pre-symptomatic stages.
- While KD was recently added to the RUSP, only 12 states have an active KD newborn screening (NBS) program.
- It is uncertain at what rate states will conform to the RUSP recommendations, with a frequently cited barrier being the absence of studies addressing the impact of HSCT on quality-of-life.
- The purpose of this study was to investigate patient/parent reported outcomes of HSCT by comparing qualitative and quantitative data collected during structured caregiver interviews.

Methods

- 90-minute caregiver interview was designed to evaluate patient/family-centered outcomes of HSCT in relation to quality-of-life.
- Comparisons were made between 1) children not transplanted or transplanted after symptom onset **against** 2) those transplanted before symptom onset.
- Infantile KD (IKD; onset <12 months) and late infantile KD (LIKD; onset 13-36 months) were analyzed independently.
- Quantitative data was collected using the validated Leukodystrophy quality-of-life assessment (LQLA).
- The LQLA consists of 43 questions divided into 4 domains. Non-paired t-test were used to compare total score and sub-scores for each domain for non/late-transplanted versus early transplanted patients.
- Quantitative data was collected using open-ended data with common themes identified using two independent coders.

Results

Figure 1. Caregiver physical burdens

Figure 2. Caregiver emotional and social burden

Discussion

- This study provides convincing evidence that HSCT improves quality-of-life and reduces caregiver burden in IKD and LIKD.
- In general, early transplanted families benefitted from improved familial and social relationships, less missed educational opportunities, less financial strain, better sleep, and less time providing care. They also felt a greater sense of control over their life.
- Although there was variability between individual responses, parents of early transplanted patients often suffered less physical stress.
- This data will be invaluable to states debating whether to add KD to their NBS panels with hopes to facilitate early diagnosis and treatment.
- It will also allow families to weigh the risks and benefits of HSCT more confidently when making the life-altering decision of whether to proceed with transplant.

FUNDING: We are immensely grateful to the Rosenau Family Research Foundation for providing the funding for this project

References

Nicholas Bascou presented an abstract poster at the 2025 WORLDSymposium in February for findings resulting from research done with support from a Rosenau Family Research Foundation grant awarded to KrabbeConnect titled "Parent-Reported Disease Burden in Infantile and Late-Infantile Krabbe Disease: A comparison of QOL in Transplant and Non-or-Late Transplant Patients."

View the Poster

Grant Research Findings

Susanna McColley, MD and her team at Lurie Children's Hospital have published their manuscript, "The Newborn Screening Experience of Caregivers of Children With Cystic Fibrosis in the United States: A Cross-Sectional Survey", in Pediatric Pulmonology. Findings in this manuscript come with support from a Rosenau Family Research Foundation grant titled "Caregiver and Clinician AwaREness for early Cystic Fibrosis diagnosis (C-CARE CF)". Additionally, they have published a newborn screening resource page on the Lurie Children's website. You can learn more about this grant, and Susanna McColley's work, on the RFRF website.

Read the Manuscript

Newborn Screening Resources

A landmark article published earlier this spring by **Philip Farrell, MD, PhD** and team in the International Journal of Neonatal Screening, outlines the new Cystic Fibrosis Foundation-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 (TLOAF), 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 Article

Another article published by **Philip Farrell, MD, PhD** and team in Pediatric Pulmonology this past spring was based on the whole genome sequencing studies completed with TLOAF/RFRF funding on the first cohort of children with Cystic Fibrosis who were diagnosed through screening.

Read the Article

Connect With Us!

General Information

www.rosenaufoundation.org
info@rosenaufoundation.org

Grants

rosenaufoundation.org/grants-process/
grants@rosenaufoundation.org

@RosenauFoundation
@rosenaufoundation
@rosenaufoundation
@Rosenau_Fdn