

A LETTER FROM THE PRESIDENT

In 2024, Global DARE Foundation made remarkable strides in advancing its mission. One of the year's important achievements was the release of updated dietary guidance, built upon new food testing results from 2023. Recognizing the critical role of diet in managing Refsum Disease, this update introduced a two-level dietary guidance system, similar to those used in other metabolic conditions. This system empowers patients with clearer, more actionable information, enabling them to make informed dietary choices to manage their phytanic acid levels more effectively.



Our presence at the Association of Research in Vision and Ophthalmology (ARVO) conference and the Foundation Fighting Blindness conference in the US significantly raised awareness of Refsum Disease among medical professionals, researchers, and patients, broadening the reach of our advocacy efforts.

During October's Refsum Awareness Week, we hosted four impactful webinars covering essential topics such as navigating mental health, updates on Refsum clinical care, updates on Refsum research, and a Global DARE update. These sessions provided valuable insights and practical advice to the community. Beyond the webinars, we highlighted new patient stories, showcasing the lived experiences of individuals affected by Refsum Disease and emphasizing the strength and resilience within the community. Our Country Ambassadors further amplified these efforts, driving awareness and advocacy in their respective regions.

A defining moment for Global DARE in 2024 was receiving the prestigious \$800K Chan Zuckerberg Initiative Rare as One grant. This transformative funding will significantly enhance our work over the next five years, empowering us to advance research, strengthen community engagement, and improve resources for those impacted by Refsum Disease.

This year also marked our 5-year anniversary, a milestone reflecting significant achievements in raising awareness, improving dietary guidance, expanding disease understanding, building community, and advancing research. None of this would have been possible without the amazing support of our community. Together, we continue to drive meaningful change and a brighter future for all those living with Refsum Disease.

Kristie DeMarco | President
Global DARE Foundation

Refsum Awareness Week - October 12-19

ONE (1) AMAZING WEEK

Four Not-to-be-Missed Webinars

Navigating
Mental
Health

Clinical
Guidelines
Overview

Advances in
Refsum
Research

Celebrating
5 Years of
Progress

Presented by the Global DARE Foundation

BOARD OF DIRECTORS

In 2024, the Global DARE Foundation Board of Directors continued to provide strategic leadership, advancing initiatives aligned with our mission to improve care, increase research, raise awareness, build community, and ensure long-term sustainability and growth. The Board remains steadfast in driving impactful programs that enhance the quality of life for individuals affected by Refsum Disease.



Kristie DeMarco
President



Susan Kuranoff
Vice President



John DeMarco
Treasurer



Marissa Rossouw
Secretary



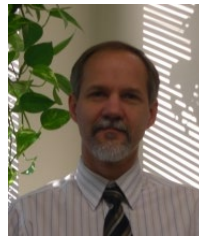
Darrell Gunter
Director



Mark Liquorish
Director



Heidi Page
Director



Donald Samulack
Director



Kerstin Reichert
Director

As part of Refsum Awareness Week 2024, Global DARE Foundation hosted a special webinar on October 19th to commemorate the Foundation's 5th anniversary. Board members Kristie DeMarco and Susan Kuranoff reflected on the Foundation's transformative accomplishments over the past five years, highlighting key initiatives such as the release of updated dietary guidance, the establishment of the Boston Refsum Center of Excellence, and the expansion of awareness programs.

Looking ahead, the webinar unveiled the Foundation's new 5-year strategic plan, focusing on strengthening research networks, advancing patient care, fostering community engagement, and ensuring operational sustainability. The event included a dynamic Q&A session where Kristie and Susan addressed audience questions, offering unique insights into the Foundation's work and sharing stories from their personal journeys with Refsum Disease.

The full webinar is available for viewing at this link: [Celebrating 5 Years of Success and a Vision for the Future](#). We encourage everyone to watch and learn more about our mission and the incredible progress we are making together.

MEDICAL & SCIENTIFIC ADVISORY BOARD

The Global DARE Foundation's Medical & Scientific Advisory Board (MSAB) is comprised of leading researchers and clinicians in the fields of Refsum Disease, peroxisomal disorders, and Retinitis Pigmentosa. Their expertise is integral to guiding the Foundation's initiatives, ensuring our efforts in improving care, advancing research, and raising awareness are grounded in scientific rigor and clinical excellence.

In 2024, the MSAB played a key role in two impactful webinars. The first, on Refsum Disease Clinical Care, featured Dr. Radha Ramachandran, MD, PhD, who provided an overview of the first-ever clinical guidelines for Adult Refsum Disease. These guidelines are a significant step forward, offering essential care frameworks to clinicians for improving quality of life for patients. Link: [Adult Refsum Disease - Clinical Guidelines Overview](#)

The second webinar focused on research advancements, with Dr. Wedad Fallatah presenting the ongoing Refsum mouse model study at Kennedy Krieger Institute. This study is providing valuable data to inform future research. Dr. Rachel Huckfeldt then discussed gene-agnostic clinical trials for Retinitis Pigmentosa, a key concern for individuals with Refsum Disease. These updates highlight the Foundation's commitment to cutting-edge research and its potential to improve patient outcomes. Link: [Advances in Refsum Disease Research](#)

Through the continued dedication of the MSAB, Global DARE remains at the forefront of progress in Refsum Disease research and care.



Joe Hacia, PhD
Chair



Paul Watkins,
MD, PhD,
Co-chair



Mousumi
Bose, PhD
Co-Chair



Ronald
Wanders, PhD



Florian
Eichler, MD



Radha
Ramachandran,
MD, PhD



Anthony
Wierzbicki, PhD



Sarah Firman
Dietitian



Rachel
Huckfeldt, MD,
PhD



Ann Moser,
PhD



Sacha
Ferdinandussa,
PhD



Bart Leroy,
MD, PhD



Wedad Fallatah,
MD, PhD



Reinhard
Klingel, MD,
PhD

COUNTRY AMBASSADOR PROGRAM

The Country Ambassador program is a cornerstone of the Global DARE Foundation's mission to raise awareness of Refsum Disease worldwide. Ambassadors work tirelessly to establish and strengthen relationships within their countries, fostering awareness and providing support to individuals affected by Refsum Disease. As official liaisons between their country and Global DARE Foundation, Ambassadors are active in driving our efforts to promote earlier diagnoses and connect individuals with the global Refsum community.



Susan Kuranoff,
Switzerland



Kristie DeMarco,
United States



Alan Gunzburg,
United States



Mark Liquorish,
United Kingdom



Kerstin Reichert,
Germany



Bob Wyborn,
Australia



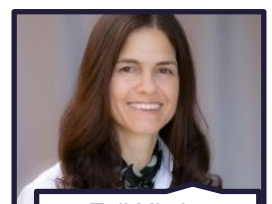
Susie Breuer,
Australia



Sharon Burstin,
Australia



Michel Veys,
Belgium



Tali Mirsky
Lachman, Israel

Key Accomplishments

Welcoming a New Ambassador: This year, we welcomed Tali Mirsky Lachman as our Country Ambassador for Israel. Inspired by her 21-year-old son Guy's diagnosis of Refsum Disease in 2023, Tali and Guy are passionately committed to expanding the network of patients, medical professionals, and researchers. Together, they aim to raise awareness and accelerate progress in research and therapy development.

Raising Awareness Worldwide:

- **Australia:** Sharon Burstin partnered with the Centre of Eye Research to publish her case report, highlighting the profound challenges of lengthy diagnostic journeys for people with Refsum Disease.
- **Switzerland:** Susan Kuranoff collaborated with Navigating Blindness to share her personal story, which was featured on their social media channels to raise awareness globally.
- **Germany:** Kerstin Reichert founded Refsum Deutschland e.V., a national organization aimed at raising the profile of Refsum Disease and uniting the community in Germany.
- **United States:** Kristie DeMarco, serving as the Foundation Fighting Blindness Maine Chapter President, collaborated with other chapter leaders during the annual meeting to raise awareness of Refsum Disease across multiple states.

Through their unwavering dedication, our Country Ambassadors continue to inspire change, empower patients, and strengthen the global network of individuals and organizations committed to tackling Refsum Disease.

REFSUM DEUTSCHLAND E.V. – 2024 HIGHLIGHTS

Global DARE Foundation is proud to collaborate closely with Refsum Deutschland e.V., a vital partner in raising awareness and advancing the path toward a cure for Refsum Disease. Founded in 2023, Refsum Deutschland e.V. has grown to 43 members, including patients, families, and advocates, and continues to champion support and resources for the Refsum community in Germany.

In 2024, the organization achieved key milestones, including launching a dedicated homepage and flyer to provide essential information to patients, families, and medical professionals. Translations of the Global DARE Foundation's dietary video and guide into German ensured critical resources are more accessible.

Additionally, Refsum Deutschland e.V. expanded its network by connecting with new patients, hosted two online exchange sessions, and played a pivotal role in adding two experts to the Global DARE Foundation Medical Advisory Board. Initial discussions to establish Germany's first competence center for Refsum Disease at the University Hospital in Leipzig also marked a major step forward.



REFSUM AWARENESS WEEK

Global DARE Foundation hosted its fourth Refsum Awareness Week from October 12–19, bringing the Refsum community together through a series of virtual events aimed at raising awareness and promoting early detection and diagnosis. The week featured four live webinars covering vital topics: mental health strategies for those managing chronic illness, updates on Refsum related research, the latest clinical guidelines for Adult Refsum Disease, and a special session commemorating Global DARE's 5th anniversary with reflections from Kristie DeMarco and Susan Kuranoff. Link: [Refsum Awareness Week Videos](#)

A virtual 5K activity encouraged participants to run, walk, bike, swim, or roll throughout the week, helping raise funds and awareness for Refsum Disease. With over \$22,000 raised, this event demonstrated the Refsum community's dedication to advancing our mission of improving the lives of those with Refsum Disease. Refsum Awareness Week 2024 highlighted the importance of collaboration in advancing research and improving care for those affected by Refsum Disease.

FOOD TESTING PROGRAM

Our food testing program continues to be a cornerstone of support for the Refsum Disease community. This program provides essential information about the phytanic acid content in various foods, empowering individuals to make informed dietary choices and effectively manage their condition.

In 2022, the Refsum Diet guidelines were fully reformatted to integrate the latest food testing results funded by the Global DARE Foundation. The updated guidelines use a traffic light system to categorize foods:

GO	CAUTION	STOP
Foods that can be eaten freely	Foods to be eaten in limited quantities	Foods which should be avoided entirely

Initially, the guidelines assumed that phytol fatty esters and free phytol—primarily found in vegetables and legumes—are absorbed in the gut and readily convert to phytanic acid in humans. This assumption led to several high-phytol fatty ester foods being classified as “Stop.”

In 2024 the guidelines were further revised to incorporate results of additional food testing done in 2023. This update retains the traffic light system but introduces Level 1 and Level 2 recommendations:

- Level 1 advice focuses solely on the phytanic acid content of foods. This update resulted in reclassification of certain items and added broader dietary recommendations, including guidance on regular meals, carbohydrate intake, exercise, and illness management.
- Level 2 advice addresses foods high in phytol fatty acid esters, which may potentially influence phytanic acid blood levels. Since current food analysis cannot conclusively determine free phytol content, free phytol has been excluded from Level 2 recommendations until further research provides clarity.

Refsum dieticians advise that all individuals with Adult Refsum Disease should follow Level 1 dietary recommendations as a baseline. If phytanic acid levels remain high despite adhering to Level 1 advice, your diet should be reviewed for Level 2 foods, as these could be contributing to elevated levels. It is essential to consult a local dietician before making any significant changes to regular eating habits.

The updated guidelines were presented by the Refsum dieticians in a webinar held in January. Link: [Presentation on the 2024 Updated Refsum Diet Guide](#)

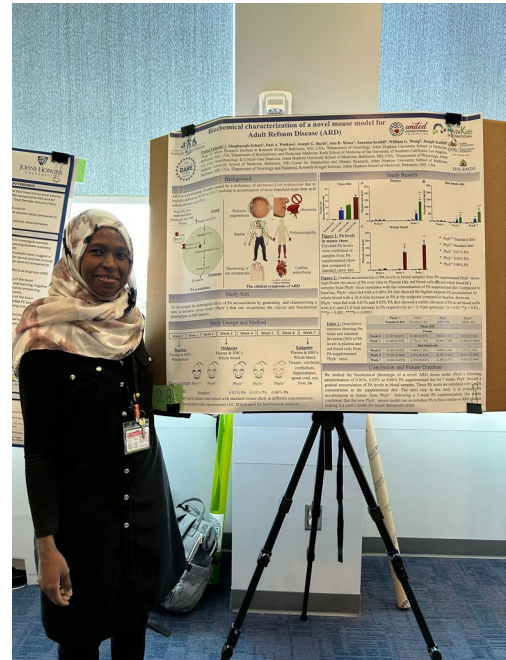
The program’s continuation is only possible thanks to the generous support of our community, whose contributions directly impact the lives of those we serve.

PROGRESS ON REFSUM DISEASE RESEARCH

During Refsum Awareness week, Dr. Wedad Fallatah from the Kennedy Krieger Institute (KKI) shared exciting progress in the research on a novel mouse model for Adult Refsum Disease, developed in collaboration with research teams at the Johns Hopkins School of Medicine and KKI. This mouse model mimics the pathogenic genetic variants seen in patients with Adult Refsum Disease, opening new opportunities to better understand disease progression and test potential therapies.

Study Highlights:

The research focused on understanding how the accumulation of phytanic acid, a key feature of Adult Refsum Disease, affects the body, especially in the blood and tissues. Dr. Fallatah and her team studied four groups of mice, each on a specific diet: a standard diet and diets supplemented with varying levels of phytanic acid. Over a seven-week study, blood samples revealed a gradual and significant increase in phytanic acid levels in the supplemented groups, particularly in the highest concentration diet group.



These findings suggest the mouse model could be a valuable tool for studying phytanic acid toxicity in Adult Refsum Disease and potentially testing therapeutic strategies. Ongoing research is now focusing on tissue analysis, especially the brain and eye, to begin to evaluate how phytanic acid accumulation impacts tissue health and function.

This groundbreaking study was partially funded by a \$60,000 grant from the Global DARE Foundation, made possible by our generous community of supporters.

Recognition of Excellence:

Dr. Fallatah's outstanding work was recently honored with the Best Poster Award in the Biomedical Research category at the 10th Annual Johns Hopkins Postdoctoral Conference.



Many thanks to the generous support of the research team, Drs. Joey Scafidi, Paul Watkins William Wong, Joseph Hacia (University of Southern California, and Prof. Ann Moser who helped to make this research project possible.

REFSUM PATIENT REGISTRY

In August of 2021 Global DARE Foundation rolled out the first ever patient registry. A patient registry brings together the Refsum community and researchers to gain a better understanding of Refsum Disease. We continue to learn from the data being collected in the registry. Below are some of metrics collected to date.

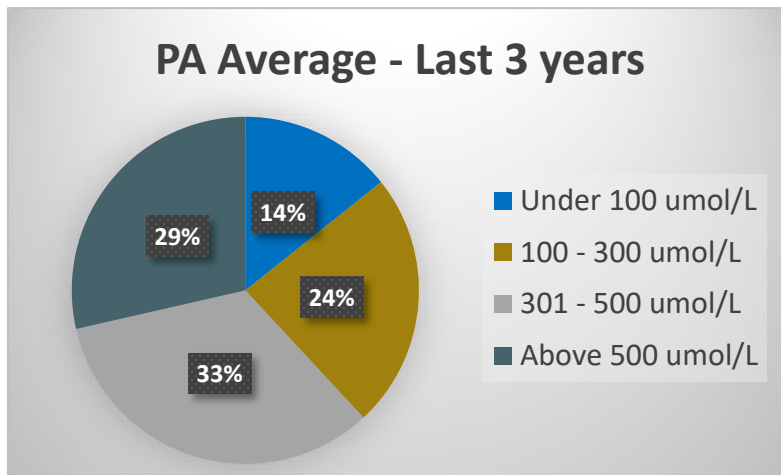
45 participants
(33 sharing data
with DARE)

13 Countries
Represented

Average 16
years to
diagnosis

43% use a
visual aid

38% have an
assistive
hearing device



Current Symptoms	%
Retinitis Pigmentosa	97%
Anosmia	79%
Shortened Toes/Fingers	76%
Peripheral Neuropathy	69%
Hearing Loss	59%
Ataxia	55%
Other Bone Abnormalities	52%
Ichthyosis	34%
Cardiac	14%

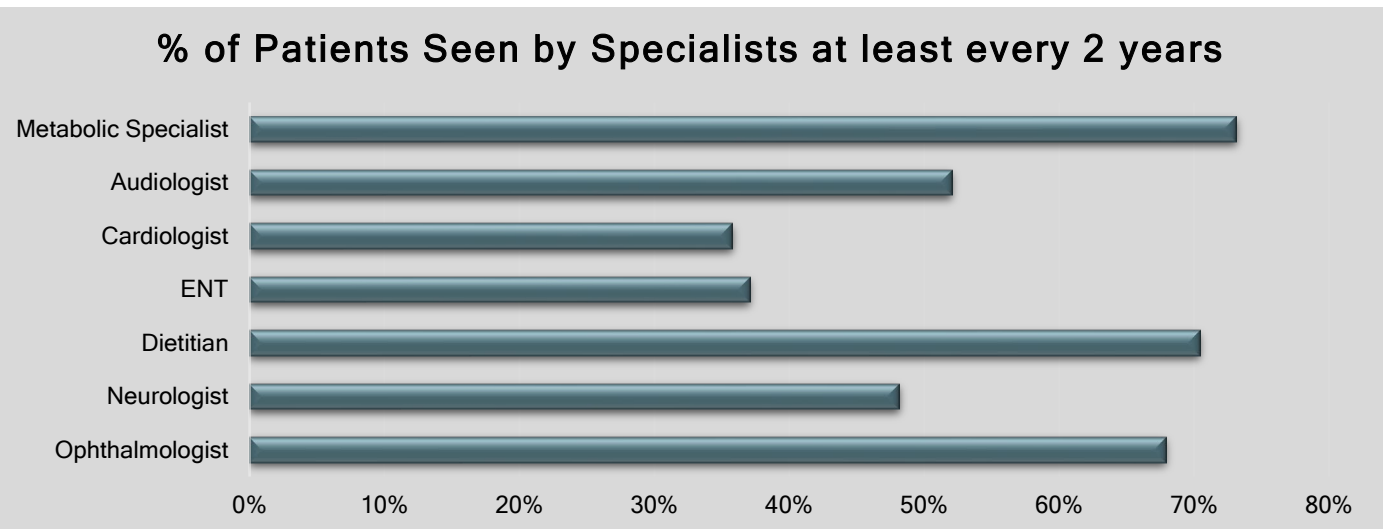
24% have had
Plasmapheresis

69% Diagnosed
through Genetic
Testing

59% have had
hospitalizations
due to Refsum

73% have daily
pain

59% had to
change exercise
due to Refsum



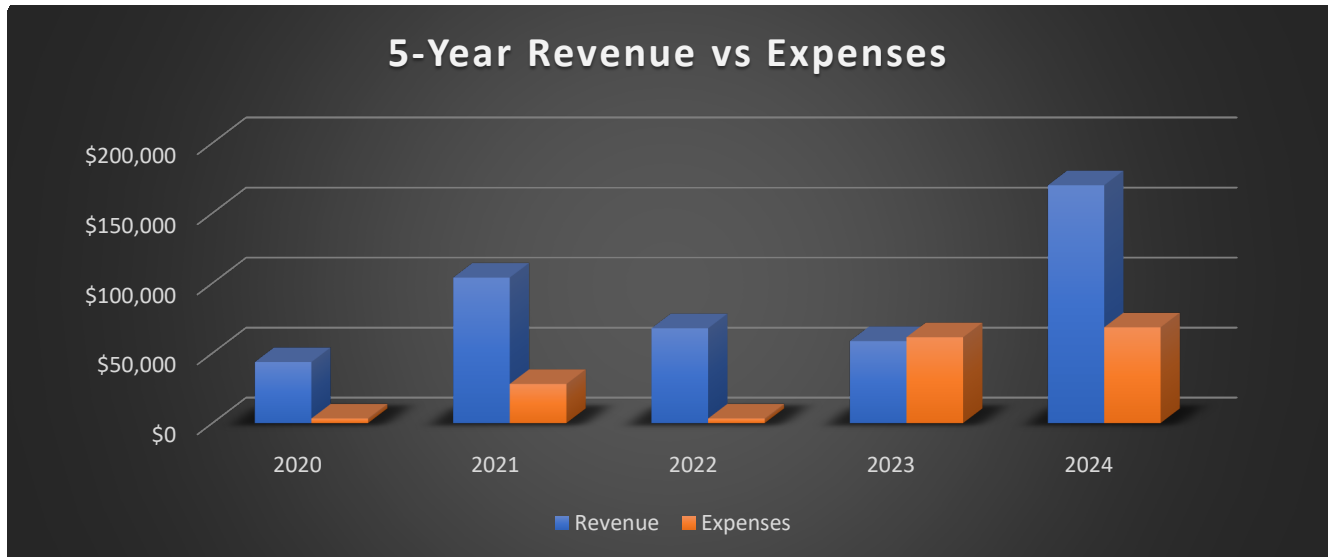
¹ Null values are not included as part of the percentages shown in this presentation

² 4 of the 45 are carriers of Refsum. Carriers are not included in the metrics above

You can learn more about the [Refsum Patient Registry](#) at our website.

5-YEAR FINANCIAL UPDATE

Over the past five years, the Global DARE Foundation has demonstrated steady financial growth and a strong commitment to fiscal responsibility. As illustrated in the year-over-year revenue vs. expense graph, we have successfully increased our revenue while keeping operating expenses low, ensuring that our resources are effectively allocated to support our mission.



¹2024 is estimated

In 2024, we set an ambitious fundraising goal of \$93,000, and we far exceeded it at an estimate of \$170,000! We were honored to be awarded the prestigious \$800,000 CZI Rare as One five-year grant, with \$100,000 received in 2024, providing a transformative boost to our mission.

We are proud to share that 78% of our expenses over this 5-year period has directly funded our three key programs: expanding global awareness of Refsum Disease, supporting our vital food testing program, and advancing critical research. These achievements wouldn't be possible without the tremendous support from our community, whose generosity and commitment continue to drive us forward.

Thank you for helping us make a lasting impact on the lives of individuals and families affected by Refsum Disease.