

2023 ANNUAL REPORT

Global



#CureRefsum



Global DARE Foundation
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A LETTER FROM THE PRESIDENT

As we bid farewell to another remarkable year, Global DARE Foundation is thrilled to reflect on the impactful milestones achieved in 2023. Our commitment to raising awareness and supporting the Refsum community has never been more evident, and we are grateful for the incredible strides made together.



This year we partnered with both Foundation Fighting Blindness and United Leukodystrophy Foundation to host conference booths at their events. These booths served as a dynamic platform to raise awareness about Refsum disease. The engaging discussions and informative materials helped us connect with a diverse audience, fostering a deeper understanding of Refsum and its impact on individuals and families.

The Refsum Awareness Week was another triumph, marked by the powerful narratives shared during the week-long campaign. Patient stories took center stage, providing a real-world glimpse into the challenges faced by those living with Refsum. This initiative not only elevated awareness but also underscored the importance of empathy and support within our community.

In a groundbreaking move, Global DARE proudly awarded its first-ever \$60,000 grant, a pivotal step in advancing research and support for individuals affected by Refsum. This significant investment is a testament to our dedication to making a tangible difference in the lives of those living with Refsum.

In our mission to build a stronger community, Global DARE hosted informal Refsum socials in both London and Scotland, bringing individuals together to share experiences, forge connections, and offer mutual support. These gatherings were instrumental in fostering a sense of belonging and solidarity among those touched by Refsum.



Further reinforcing our commitment to building research enabling capabilities, the results of the Refsum patient registry data were published in May of 2023. This invaluable resource, made possible by Global DARE, provides crucial insights that will shape future research and enhance the quality of care for individuals with Refsum.

As we celebrate these achievements, we extend our gratitude to the dedicated supporters, volunteers, and partners who have made this journey possible. Global DARE looks forward to continuing our mission in the coming year, united in our commitment to making a positive impact on the lives of those affected by Refsum disease.

Kristie DeMarco | President
Global DARE Foundation

BOARD OF DIRECTORS

In 2023 the Global DARE Foundation Board of Directors continued with the 3-year strategic plan established in 2022. The plan focused on driving initiatives to improve care, increase research, raise awareness, build community and create long term stability sustainability and growth. Below you will find the goals established to guide these initiatives.

Research & Care

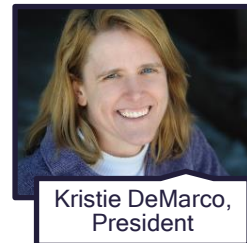
- Improve treatment and care by establishing a network of centers around the world with Refsum experience
- Execute on the 3-year research roadmap including the food testing program, research studies, and building research enabling capabilities
- Increase participation in the patient registry to 75%
- Enable better access to Refsum research papers

Awareness & Community

- Reduce the time to diagnosis through education and awareness
- Conduct yearly surveys of the Refsum community to understand the best ways to achieve our mission
- Engage the community in webinars and in-person conferences
- Expand the Country Ambassador program to raise awareness around the world
- Increasing the number of patients connected with DARE.
- Establish an annual Refsum Disease Awareness Week

Sustainability & Growth

- Build and execute strategic plans across all areas of focus tied to the DARE mission
- Ongoing education and development of the newly expanded Board of Directors
- Determine the necessary data, tools, and technology needed for long term sustainability
- Increase annual fundraising by 15% per year.
- Pursue grant opportunities through various channels



Kristie DeMarco,
President



Susan Kuranoff,
Vice President



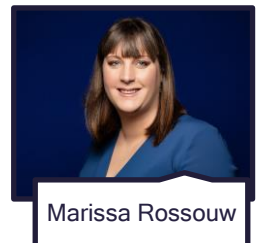
John DeMarco,
Treasurer



Kim Bruce,
Secretary



Mark Liquorish



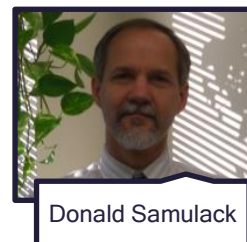
Marissa Rossouw



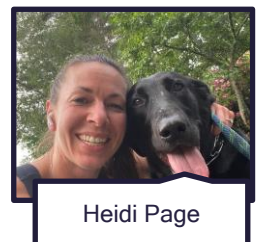
Zahra Dhanani



Darrell Gunter



Donald Samulack



Heidi Page

As you will see from the rest of the Annual report, we continue to make great progress in the second year of the strategic plan.

MEDICAL & SCIENTIFIC ADVISORY BOARD

Our Medical & Scientific Advisory Board (MSAB) is made up of the foremost researchers and clinicians in the field of Refsum Disease, peroxisomal disorders and Retinitis Pigmentosa.

The MSAB is constantly collaborating with us on how to improve treatment and care and direct the areas of focus for Refsum research.

Research Grant Award

With the support of our MSAB, Global DARE Foundation approved the funding of \$60,000 towards an adult Refsum Disease (ARD) mouse model study that will be conducted at Kennedy Krieger Institute.

The goal of the study is to generate genetically engineered PHYH-mutant mice that accumulate phytanic acid when placed on special diets and determine if they develop ARD symptoms, such as progressive vision and hearing loss and peripheral neuropathy.

If successful, the ARD mouse model can be used to better understand why phytanic acid is toxic in people and test therapies aimed at reducing phytanic acid toxicity in order to improve the lives of people living with ARD.

The preliminary data generated from this study can be used to apply for larger federal grants to perform more comprehensive studies aimed at finding better treatments for ARD.

Other Initiatives

The Medical and Scientific Advisory Board is also actively working on clinical guidelines and updated dietary guidance which will provide significant benefits to people with Refsum Disease in the short term.



Joe Hacia, PhD
MSAB Chair



Paul Watkins, MD,
PhD, MSAB co-chair



Mousumi Bose, PhD
MSAB co-chair



Florian Eichler, MD



Ronald Wanders,
PhD



Radha
Ramachandran, MD,
PhD



Bart Leroy, MD, PhD



Rachel Huckfeldt,
MD, PhD



Eleanor Baldwin
Dietician



Anthony Wierzbicki,
PhD



Sacha
Ferdinandusse, PhD



Sarah Firman,
Dietician



Ann Moser, BA



Wedad Fallatah, MD,
PhD

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. In May 2023, the results of this Refsum patient registry data were published making the data available for use amongst the research community. This provides crucial insights that will shape future research and enhance the quality of care for individuals with Refsum. [Read the research paper](#) below to learn more.

Phytanic Acid Intake and Lifestyle Modifications on Quality of Life in Individuals with Adult Refsum Disease: A Retrospective Survey Analysis

by  Jeffrey J. Li * ,  Jane J. Kim and  Fauzia Nausheen

Department of Education, California University of Science and Medicine, Colton, CA 92324, USA

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Nutrients **2023**, *15*(11), 2551; <https://doi.org/10.3390/nu15112551>

Submission received: 10 May 2023 / Revised: 26 May 2023 / Accepted: 27 May 2023 / Published: 30 May 2023

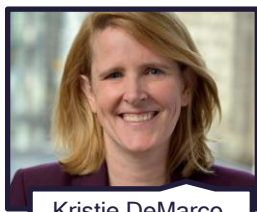
You can learn more about the [Refsum Patient Registry](#) at our website.

COUNTRY AMBASSADOR PROGRAM

Our Country Ambassador program continues to raise awareness of Refsum disease around the world and is vital to our mission. In 2023 we welcomed a new Country Ambassador, Kerstin Reichert in Germany. Country Ambassadors are responsible for establishing and building relationships within their country to increase awareness and support for those with Refsum disease. Ambassadors serve as official liaisons between their country and the Global DARE Foundation and are essential in achieving our mission. Our hope is that the program will help people with Refsum receive an earlier diagnosis and connect people who have already been diagnosed with the Refsum community.



Susan Kuranoff,
Switzerland



Kristie DeMarco,
United States



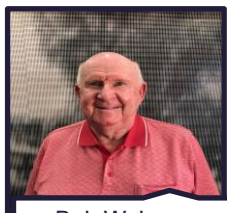
Alan Gunzburg,
United States



Mark Liquorish,
United Kingdom



Kerstin Reichert,
Germany



Bob Wyborn,
Australia



Susie Breuer,
Australia



Sharon Burstin,
Australia



Michel Veys,
Belgium

DRIVING EDUCATION AND AWARENESS

As part of Global DARE Foundation's goals, we look to raise awareness and conduct educational webinars and videos to all that are interested in learning more about Refsum Disease

DARE Fest 2023: This year we held our second annual DARE Fest, with the objective of continuing to increase awareness of Refsum and the resources available to people with Refsum and the medical community.

At DARE Fest 2023 we hosted a variety of exciting virtual events to educate, raise awareness, build community around the world and have some fun. For the first time we held a free Mindfulness Practice Webinar to kick off DARE Fest. [Click on the DARE Fest logo to the right to watch all the great content.](#)



Our DARE Fest 5K returned again this year. This virtual 5K activity (run, walk, bike, swim, etc) took place throughout the week. Additionally, we added a Diet Challenge, where individuals could participate in a challenge to follow the Refsum diet for 5, 7 or 9 days. Through these events and as part of DARE Fest, we raised \$30K USD.

Refsum Breakout Session at the ULF conference

Global DARE Foundation hosted the first hybrid Refsum Breakout session at the ULF conference in Chicago on June 23rd. The session was attended by patients, researchers and medical professionals.

The session provided an overview of Refsum Disease & Global DARE Foundation, and an update on Refsum Research. In addition, several of our Medical and Scientific Advisory Board Members introduced themselves and provided their backgrounds and interest in Refsum Disease. Watch the recorded session below.



FOOD TESTING PROGRAM

The ongoing food testing program will be taking a different approach moving forward. This program, supported by the Global DARE Foundation, has been crucial in helping people with Refsum Disease make informed dietary choices. Current testing has not only looked at phytanic acid but also its precursors (building blocks) like phytol and phytol fatty esters. These esters were added to the testing because of a study by Krauss et al. in 2017. It suggested that when we eat foods with these esters, our bodies release something called trans-phytol during digestion. This trans-phytol then turns into phytanic acid through a process called oxidation. This means that foods high in phytol fatty esters could significantly add to the daily phytanic acid intake.

Krauss's research was done in a lab, which means it doesn't tell us how well people actually convert phytol fatty esters into phytanic acid. Tests at the Kennedy Krieger Laboratory, where we've been doing our food testing, have shown that some foods have lots of phytol fatty esters. Because of this, they've been put either in the "caution" or "stop" categories in the diet, meaning people should eat them in limited quantities or avoid them all together. But we're not entirely sure if these categorizations are necessary.

To get a better understanding, the Global DARE board and the Medical & Scientific Advisory Board have decided to take a more targeted approach by investing in basic research to learn more about this conversion. As a first step, we'll start testing on mice early next year. We'll also investigate other research possibilities that can help dietitians better interpret our food testing results. This will ultimately lead to more accurate dietary information for those impacted with Refsum Disease, helping us improve the lives of those affected. We thank you for your continued support to our program.

Updated Refsum Diet Plan

Global DARE Foundation, together with the Guy's & St. Thomas Dieticians will soon be releasing the updated diet plan based on our latest food testing results. This revised plan aims to provide more comprehensive dietary guidelines for individuals managing Refsum Disease.

In conjunction with this release, a webinar is scheduled for 17th January. You can [register](#) now to receive the new information. We are eager to share the update and look forward to wide participation.



**** Save the date ****
Refsum Diet update
Webinar
Wednesday, 17-January,
4-5 pm CET



FINANCIAL UPDATE

Despite a slight dip in fundraising compared to last year, Global DARE fared well, thanks to steadfast community support. The funds, though slightly less, remain substantial, affirming our commitment to advancing research, patient support, and awareness. We extend sincere thanks to our generous donors for their crucial contributions.

Year over Year Income



Note: 2021 included a \$50K special one-time grant.

PROGRAM INVESTMENT

In 2023, the majority of Global DARE's funds were allocated to research, awareness campaigns, and food testing, underscoring our commitment to advancing knowledge and supporting the community. Operating expenses were minimal, ensuring maximum impact on our core initiatives.

Operating
Expense
\$5.8K

Conferences &
Raising Awareness
\$2.3K

Food Testing &
Research
\$53.4K