

# 2022 ANNUAL REPORT

## Global



# #CureRefsum



Global DARE Foundation  
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# A LETTER FROM THE PRESIDENT

Global DARE Foundation has had another amazing year. Again in 2022 we established a set of measurable goals focused on improving treatment and care, advancing research, raising awareness, and collaborating. As you will see from our accomplishments, we have achieved everything we set out to do this year.



At the end of 2021, we held an in-person meeting to talk about establishing the first Refsum Center of Excellence in the US located at Massachusetts General Hospital and Massachusetts Eye and Ear Infirmary. We set off to establish a multidisciplinary team to improve the care of people with Refsum and Refsum like disorders. The center saw its first patient in March of 2022!

We completed the testing of 60 more foods to better inform the dietary guidance. A new traffic light system was created because of this testing. New foods were found to be safe to eat and some foods previously thought safe to eat turned out high in phytanic acid. This has had a positive impact to daily living for those with Refsum Disease.

Our Medical and Scientific Advisory Board has continued to be engaged with us this year in moving forward treatment guidelines for Refsum, creating our research roadmap and has made several grant submissions to advance translations research. Our Refsum mouse colony is now ready for studies!

We launched the first annual Refsum Awareness week called DARE Fest. It was a great success! We ran an online virtual festival with live and recorded content. 20 new educational videos and live webinars were made available for the community. During this week we also ran a virtual 5K. Through the generosity of our supporters were able to raise over \$32K. A special thanks goes to my co-workers at TD Bank who helped us receive a \$10K matching grant from TD Bank.



In September, we collaborated with United Leukodystrophy Foundation and other Leukodystrophy patient advocacy groups to raise awareness to all Leukodystrophies.

We are very grateful to have such strong supporters and volunteers helping us achieve our mission. None of this would have been possible without all of you. We are positioned well for continued success in 2023!

**Kristie DeMarco | President**  
Global DARE Foundation

# BOARD OF DIRECTORS

In 2022 the Global DARE Foundation Board of Directors established a 3-year strategic 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

As you will see from the rest of the Annual report, we have made great progress in the first 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.

In 2022 members of the MSAB presented Refsum for the first time at the conference for the Global Leukodystrophy Initiative. This is an exciting step forward for Refsum research and improving care! As part of the preparation for the conference the MSAB conducted a Heat Map assessment rating the current status of Refsum research from both a therapeutic and clinical perspective. Below are the results. A score of 1 is the lowest and 5 is the highest.

Research Area	Rating
Therapeutic Development	2
Pre-Clinical Scientific Development	3.1
Outcomes / Clinical Planning	3
Advocacy	3.2
Access to Disease Modifying Therapies	1.9
Symptomatic Care	2.8
Age, Severity and Spectrum of Disease	2.5
Access to Disease Diagnosis and Therapy Development	2.3



Florian Eichler, MD  
MSAB Chair



Ronald Wanders,  
PhD  
MSAB Co-chair



Joe Hacia, PhD  
MSAB Co-Chair



Radha  
Ramachandran, MD,  
PhD



Anthony Wierzbicki,  
PhD



Eleanor Baldwin  
Dietician



Nancy Braverman,  
MD, MS



Mousumi Bose, PhD



Sacha  
Ferdinandusse, PhD



Bart Leroy, MD, PhD



Arthur Bergen, PhD



Ann Moser, BA



Joey Scafidi, MD,  
PhD



Sarah Firman,  
Dietician



Paul Watkins, MD,  
PhD

Creation of the Heat Map has established the groundwork for the areas of focus to drive research for the years to come. Being part of this conference has already helped to raise the profile of Refsum to peak the interest of additional researchers and potential investors in driving better therapies.

## COUNTRY AMBASSADOR PROGRAM

Our Country Ambassador program was established to raise awareness of Refsum disease around the world and is vital to our mission. 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



John DeMarco,  
United States



Alan Gunzburg,  
United States



Mark Liquorish,  
United Kingdom



Liz Ebbage,  
United Kingdom



Bob Wyborn,  
Australia



Susie Breuer,  
Australia



Chantal Valkenberg,  
Netherlands



Michel Veys,  
Belgium

## RARE DISEASE DAY 2022

Rare Disease Day® takes place on the last day of February each year. The week leading up to that day Global DARE Foundation again ran a Refsum Awareness Campaign. We raised awareness for Refsum disease by sharing facts about the disease and personal stories on social media. Click below to watch all the great videos we released as part of this campaign.



### Rare Disease Day 2022

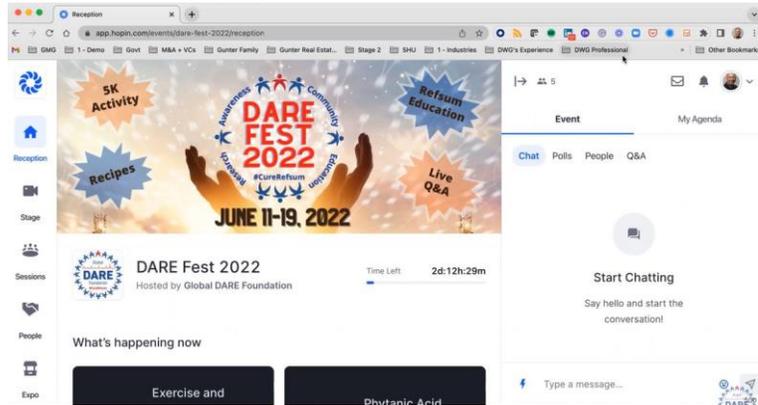


## #CureRefsum

# 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 2022:** This year we held our inaugural DARE Fest, with the objective of increasing awareness of Refsum and the resources available to people with Refsum and the medical community.

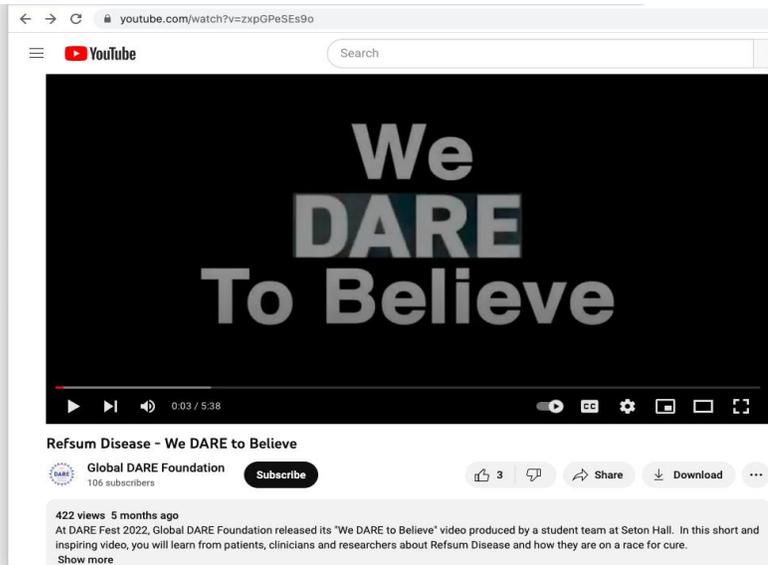


DARE Fest was an interactive week of Awareness, Community, Education and Research

comprising an Online event including presentation (recorded), live Q&A sessions, recipe competition, breakout sessions, 1:1 networking and games. These sessions were attended by patients and our medical community as a virtual conference hosted on a virtual conferencing platform called Hopin. Among the subjects explored were Phytanic Acid 101, History of Refsum Disease, Introduction to food testing, Introduction to our Centers of Excellence and discussions on areas of research plus patient, caregiver, dietician and research panels. Click on the picture above to watch all the great content.

Our Global DARE to Believe 5K returned as the DARE Fest 5K. This virtual 5K activity (run, walk, bike, swim, etc) took place throughout the week and through this event and as part of DARE Fest we raised an amount of USD \$33K.

**DARE to Believe Video:** DARE Fest also presented an ideal opportunity to launch our “We DARE to Believe” video. A five-minute video created by students at Seton Hall featuring interviews with our patients and showcasing their stories and Refsum journeys from first signs to diagnoses and into their daily lives. Click on the image to the right to watch this video.



# FOOD TESTING PROGRAM

Our program continues in collaboration with Kennedy Krieger Institute (KKI). Because diet is the only treatment available to positively influence the outcome of Refsum Disease, the DARE continues to strongly invest in our fatty acid testing program. Following the testing of 60 food items in 2021, the Refsum community identified a further 40 items for testing this year. Our current focus is on testing single ingredient foods to expand the existing repertoire to help inform people when they are reviewing ingredients on food labels. Unfortunately, food labels don't show the phytanic acid content of food; therefore, it is helpful knowing if the individual ingredients are considered safe.

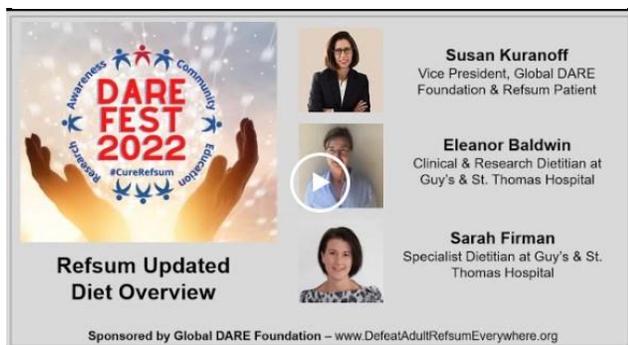
Following testing completed to date, fully revamped diet guidelines were released in June of this year. The new format presents foods according to a traffic light system

<b>GO</b>	<b>CAUTION</b>	<b>STOP</b>
Foods that can be eaten freely	Foods to be eaten in limited quantities	Foods which should be avoided altogether

While many new foods were added to the “Go” section, based on the recent knowledge that precursors to phytanic acid such as phytol and phytyl fatty esters can be converted to phytanic acid in humans, some foods have shifted to the “Caution” or “Stop” section. Regardless, it is vital for us to have an increased understanding of foods which can be safely eaten as this has a direct impact on the quality of life of people with Refsum.

To learn more about the new food guide and how the food testing is complete watch these videos.

## Refsum Update Diet Overview



**Refsum Updated Diet Overview**

Sponsored by Global DARE Foundation – [www.DefeatAdultRefsumEverywhere.org](http://www.DefeatAdultRefsumEverywhere.org)

**Susan Kuranoff**  
Vice President, Global DARE Foundation & Refsum Patient

**Eleanor Baldwin**  
Clinical & Research Dietitian at Guy's & St. Thomas Hospital

**Sarah Firman**  
Specialist Dietitian at Guy's & St. Thomas Hospital

## Food Testing Process



### MEASUREMENT OF PHYTANIC, PHYTYL FATTY ACID ESTERS AND FREE PHYTOL IN FOODS

Measured by Ann B Moser, Peroxisomal Lab, The Hugo Moser Research Institute at Kennedy Krieger Institute; Food analyses requested by the DARE Foundation, 2021 and 2022; background slides provided by Stephanie Krauss, formerly of the 

# BOSTON REFSUM CENTER OF EXCELLENCE

The first ever Refsum Center of Excellence in the United States was established this year in Boston. The multidisciplinary team provides care for children, adolescents and adults with Refsum Disease or Refsum-like disorders. They integrate state-of-the-art resources with compassionate, comprehensive care. National experts from Massachusetts General Hospital and Massachusetts Eye and Ear Infirmary work together to provide care for people with Refsum Disease. Watch the short video below by Dr. Florian Eichler to learn more about this center.

The multidisciplinary approach includes care in the areas of neurology, ophthalmology, neuromuscular, nutrition, otolaryngology (ENT), audiology, metabolism, physiatry and genetic counseling. They also work closely with all of the other departments and can make specific referrals, as needed. For the convenience of patients, a central appointment coordinator is available to schedule your appointments.



Boston Refsum Center of Excellence



## MEET THE TEAM



Florian Eichler, MD  
Neurologist



Rachel Huckfeldt, MD,  
PhD, Ophthalmologist



Reza Seyedsadjadi,  
MD, Neuromuscular



Inderneel Sahai, MD,  
Metabolic Specialist



Dominica Nichols, PhD,  
RD, LDN, Dietician



Dunia Abdul-Aziz, MD,  
ENT



Brian Wishart, DO,  
Physiatrist



Natalie Grant, Clinical  
Research Coordinator

# 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.

33 participants  
(27 sharing data  
with DARE)

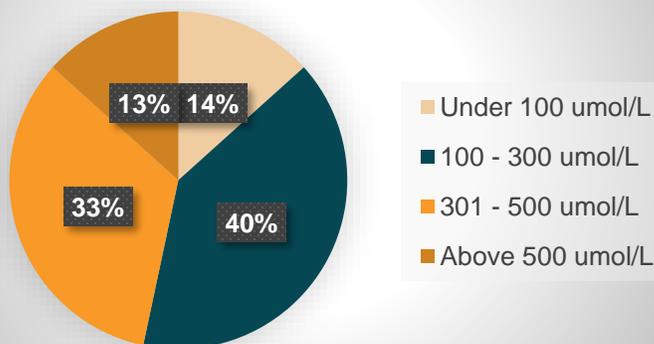
10 Countries  
Represented

Average 16  
years to  
diagnosis

33% use a  
visual aid

39% have an  
assistive  
hearing device

## PA Average - Last 3 years



Current Symptoms	%
Retinitis Pigmentosa	96%
Anosmia	78%
Shortened Toes/Fingers	74%
Peripheral Neuropathy	65%
Hearing Loss	57%
Other Bone Abnormalities	52%
Ichthyosis	48%
Ataxia	48%
Cardiac	17%

26% have had  
Plasmapheresis

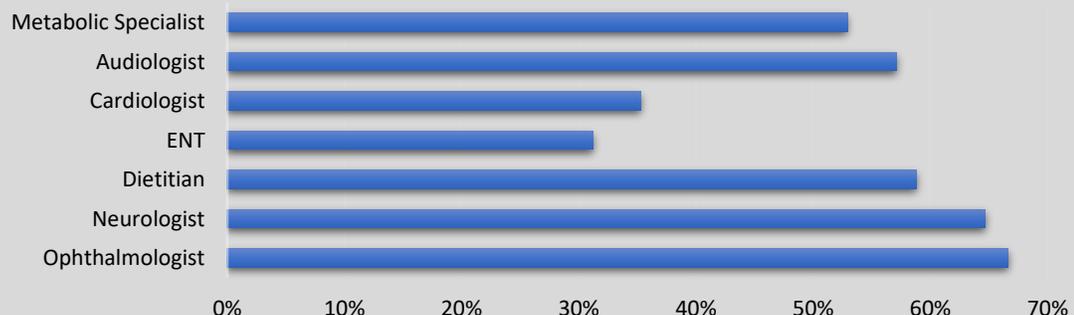
74% Diagnosed  
through Genetic  
Testing

50% have had  
hospitalizations  
due to Refsum

67% have daily  
pain

48% had to  
change exercise  
due to Refsum

## % of Patients Seen by Specialists at least every 2 years

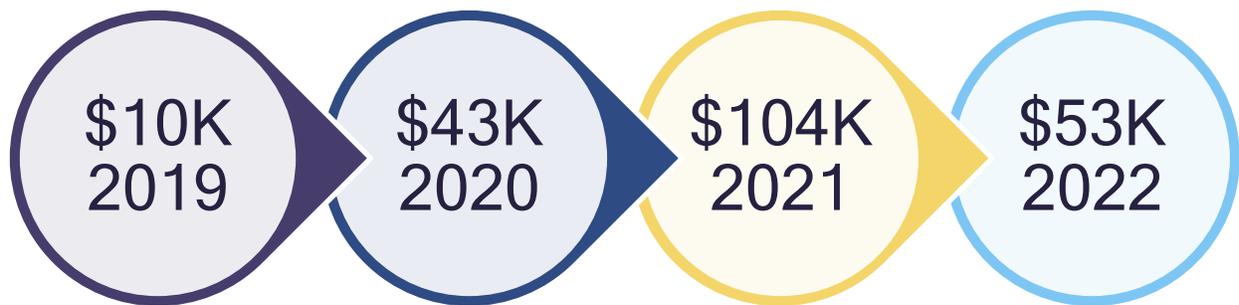


You can learn more about the [Refsum Patient Registry](#) at our website.

## FINANCIAL UPDATE

Thanks to our generous supporters, Global DARE Foundation has increased its year over year donations which has positioned us well to continue to achieve our mission of improving the lives of people diagnosed with Refsum Disease.

### Year over Year Income



Note: 2022 financials are as of November 30<sup>th</sup>. We are tracking to our goal of raising \$62,300 in 2022. 2021 included a \$50K special one-time grant.

## PROGRAM INVESTMENT

Each year we keep our expenses low and allocate greater than 90% of our funding towards our initiatives of raising awareness, food testing and research. The chart below provides a view of the budget allocation towards programs and expenses year over year.

