

FEBRUARY 2021 • VOL. 6

ROHHAD READER

GLOBAL RARE DISEASE DAY EDITION

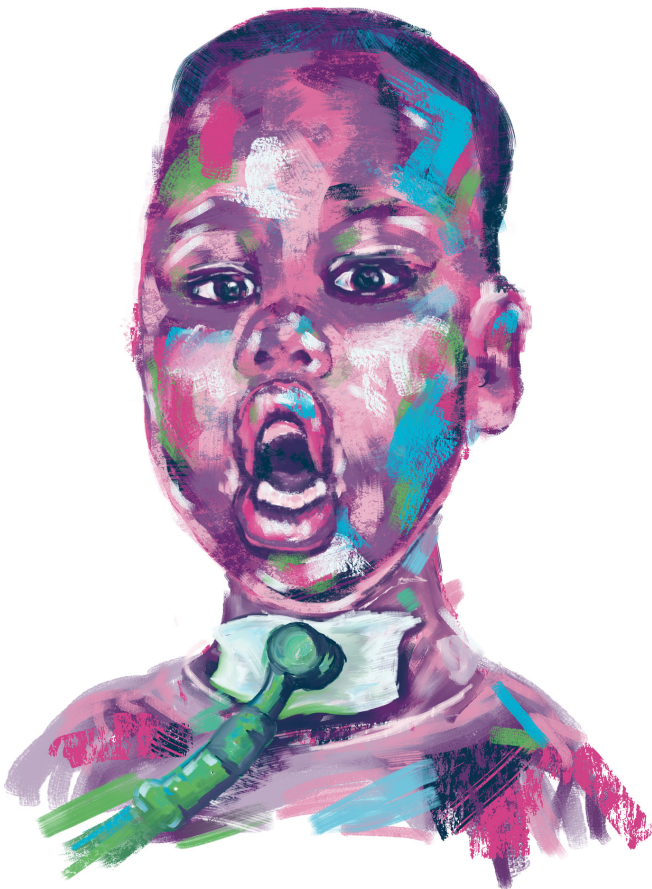


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RARE DISEASE DAY 2021
28 FEBRUARY

LEARN MORE AT
[RAREDISEASEDAY.ORG](https://rarediseaseday.org)



WELCOME BACK!

We proudly present our 6th edition of the **ROHHAD Reader**, a newsletter created for families whose daily lives are, or have been affected by, ROHHAD. Our goal is to teach and strengthen the ROHHAD community by bringing patients, families, physicians, and researchers together.

This is our special **Rare Disease Day** edition! Every year, on the last day of February, we like to take time to acknowledge the **over 300 million people living all over the world with a rare disease.**



RARE DISEASE DAY 2021
28 FEBRUARY

LEARN MORE AT
RAREDISEASEDAY.ORG



INTRODUCING...

The ROHHAD International Consortium!

Thanks to the internet and improved technology, ROHHAD families, clinicians, and researchers from all over the world have been able to connect and collaborate in ways never before possible. These various experts work hard to discover the cause(s) of ROHHAD and continue to advance care in order to provide each patient with the **best medical experience possible**.

This consortium (ROHHAD IC) is proud to be working together as one!

**Learn more about this
group on page 11!**

-The ROHHAD Reader Team



RARE DISEASE DAY®

6

CONTINENTS 

PORTRAITS 

HEROES 

LIVES 

[Link to the Rare Disease Day 2021 Official Video](#)

We are **RARE**

We are **MANY**

We are **STRONG**

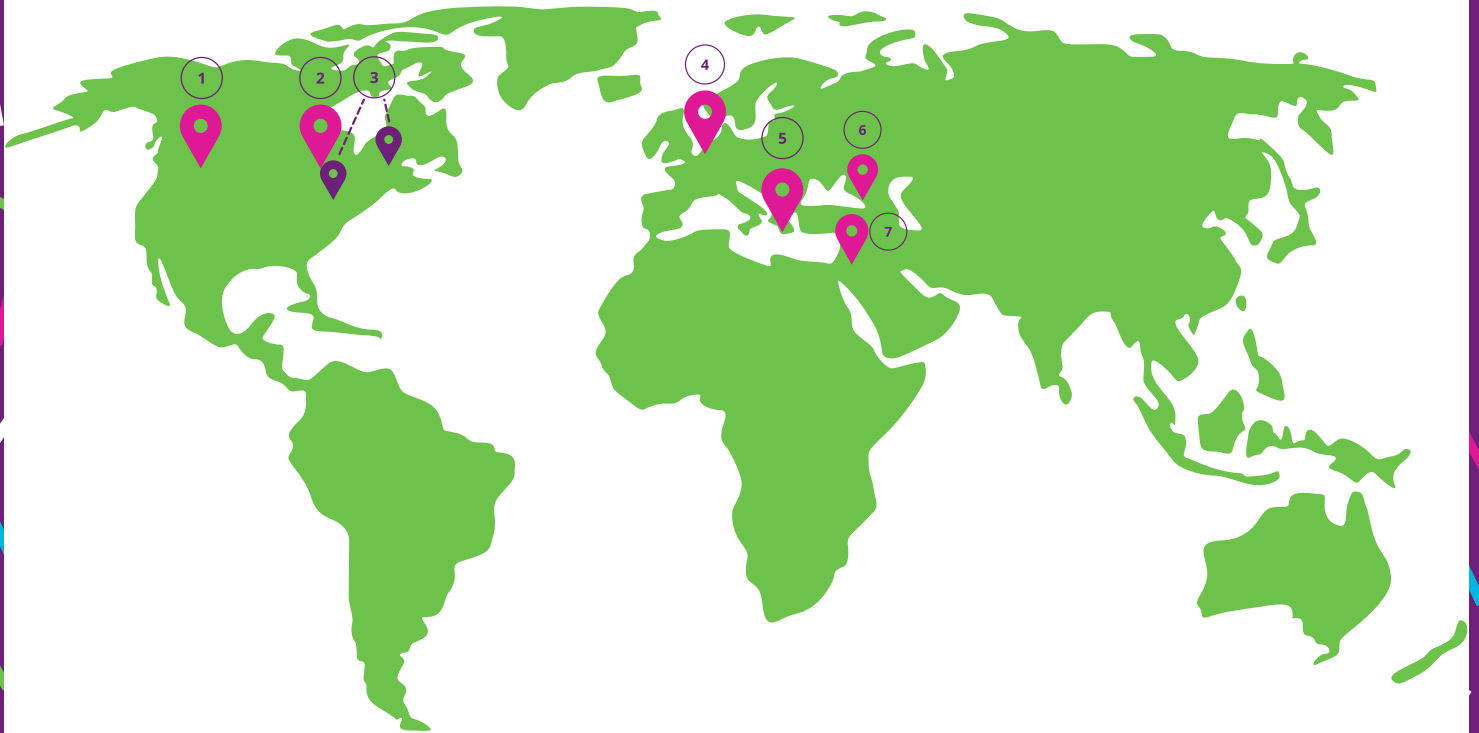
We are **PROUD**

Rare Diseases affect 300 million people world wide. It is due in part to communities like ours that these diseases get an international spotlight. That spotlight can lead to research, funding, and, ultimately, discoveries to improve the lives of everyone affected. On the next page we have an example of this with a ROHHAD Research Recap!



2020 ROHHAD RESEARCH RECAP

ROHHAD research is happening around the world. This research is meant to help doctors learn more about ROHHAD so they can find better treatments. Stay tuned for more research, and be on the lookout for studies you can participate in to help contribute to improving care in ROHHAD!



1

J. Gharial et al. (Sept 2020)

Calgary, Canada
doi.org/10.1097/mps.0000000000001927

2

S. Selvadurai et al. (Dec 2020)

Hamilton and Toronto, Canada
doi.org/10.1002/ppul.25199

3

R. Kaw and M. Kaminska (Sept 2020)

Cleveland, USA and Montreal, Canada
doi.org/10.1002/ppul.25199

4

J. Harvengt et al (July 2020)

Liège, Belgium
doi.org/10.1210/clinem/dgaa247

5

M. Filippidou et al. (Sept 2020)

Athens, Greece
doi.org/10.1210/clinem/dgaa247

6

G. Özcan et al. (Nov 2020)

Ankara, Turkey
doi.org/10.3389/fped.2020.573227

7

A.D Goldbert et al. (Oct 2020)

Beer-Sheva and Tel Aviv, Israel
Link: doi.org/10.5664/jcsm.8678

SHOW YOUR STRIPES®

ON RARE DISEASE DAY®
FEBRUARY 28TH



FIND THE RARE DISEASE DAY WORDS IN THE PUZZLE BELOW!

PWFHTEVJDKUF OFKXOEQH QKDSW
EAWARENES SY PANWURHFIVILC
XELCZMLEQDPBTQM JGSEYQLSUX
YJT TQJEYAOJFTIYQARB NHLEPC
XSCHALLENGESEGBHNIRSANAAH
WUAWMDDSEIPENAMZINUOKESUR
JPIRCWVIJZHHTCJCZTAGVSEVO
CPANQEGOSCZJIKWOAERFESYJN
BOITIPSWCOLZOFJMTRYATOOWI
TRNKRTAOMARTNDTMINWJRQLPC
JTYNGEIRVFTDRBAUOADSWEF AA
CAFVEEAATYQEENBNNTINGXNTB
NSFZJCVTTNWAGRGIRIAQGIDIU
EDNHWZTXMIEFLYCTAOGCQZAEW
UFCQBCLIKEVRDQFYFNHMEUNC
WPQJGILBOONESFNPUA OIOXPTQ
HFHIBIADINUTSFIMFLSVUEUZQ
ISNYMDKLMISDIAGNOSISCYUJZ
MSOJSPQYHVLQCVRBELSPCUWKS
PWRHHISWHKNZCZVCAREGIVERJ
AIDXEV CZDJAF CPROCLAMATION
CPYFTAUI NZCGRPMRESEARCHVK
TUJPCTLIASJAHLPZHBSOFEEZM
XMWTDUZTPNFEE OZZLXCUBSXLW
JMMJIGLRHYURVSYNDROMESAIE

ADVOCATE
ATTENTION
AWARENESS
CAREGIVER
CHALLENGES
CHRONIC
COMMUNITY
CONNECTION

DIAGNOSIS
DISEASE
DISORDER
FEBRUARY
HEALTH
ILLNESS
IMPACT

INITIATIVES
INTERNATIONAL
MISDIAGNOSIS
NORD
ORGANIZATION
PARTNERS
PATIENT

PHYSICIAN
PROCLAMATION
RARE
RESEARCH
SUPPORT
SYNDROME
TREATMENT

THIS IS A WORD SEARCH CREATED BY NORD AND RAREDISEASEDAY.ORG

COVID-19...



Has had a profound and lasting impact world-wide. New information comes to light on a regular basis, and recommendations for best practices continue to evolve. The ROHHAD Reader team wants to ensure our community members stay up to date with information appropriate for them. A few international and local resources are included below, please find and use the ones appropriate for you!

INTERNATIONAL

(INCLUDES ACCESS TO INFORMATION FOR ALL AREAS)

[International Association of Emergency Managers \(IAEM\)](#)

[World Health Organization \(WHO\)](#)

[The Global Health Network \(TGHN\) Regional Response](#)

EUROPE

[European Centers for Disease Prevention and Control \(ECDC\)](#)

USA

[Centers for Disease Control \(CDC\)](#)

AUSTRALIA

[Australian Government Department of Health](#)

VACCINES!

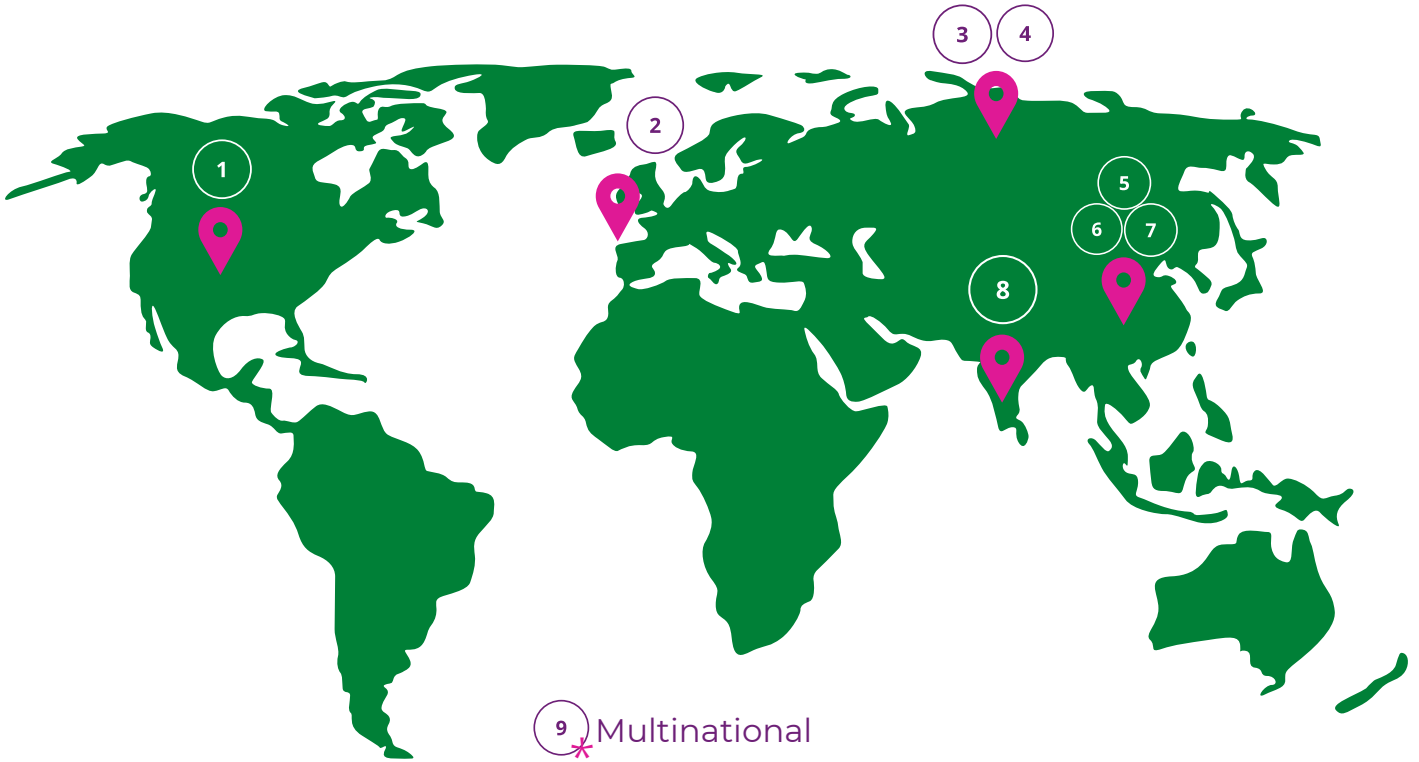


Advances in science and technology have helped create more than just the ROHHAD IC - effective vaccines against the coronavirus responsible for the COVID-19 pandemic have been developed faster than ever before! As of the beginning of February, nine different vaccines are being used across the globe. See the map on Page 8 to see where they were created!

APPROVED COVID-19 VACCINES

AS OF FEBRUARY 5TH, 2021

COUNTRY OF ORIGIN



VACCINE NAME:

1 Moderna COVID-19 Vaccine, mRNA-1273
USA

2 AstraZeneca, AZD1222
UK

3 Sputnik V
Russia

4 EpiVacCorona
Russia

5 BBIBP-CorV
China

6 Ad5-nCoV
China

7 CoronaVac
China

8 Covaxin
India

9 Comirnaty, BNT162b2
(aka Pfizer vaccine)
Multinational

More information available here:
[Regulatory Affairs Professional Society \(RAPS\)](#),
[World Health Organization \(WHO\)](#).



COVID-19 VACCINES: HEALTH EXPERTS ADDRESS THE RARE DISEASE COMMUNITY

On January 15th, 2021 the ROHHAD Reader team attended a webinar hosted by **NORD** (the National Organization for Rare Disorders) where a panel of representatives from the U.S. CDC (Centers for Disease Control) and the U.S. FDA (Food and Drug Administration) answered questions about COVID-19 vaccination.

How safe and effective are the Moderna and Pfizer vaccines for Rare Disease (RD) populations? What are the side effects for RD populations?

There weren't enough patients with RDs enrolled in clinical trials to provide a significant conclusion. **It is up to you and your doctor to discuss what is best for you.**

If I get an mRNA vaccine now, will I become ineligible for genetic therapy in the future?

Both Pfizer's and Moderna's COVID-19 vaccines are **mRNA vaccines**. **mRNA** is genetic material that contains instructions for making proteins. Once you get vaccinated, the mRNA tells the cells in your body how to make the "spike" protein found on the surface of the COVID-19 virus.

Your body will create cells with this protein and your immune system will produce **antibodies, cells that fight against viruses**. Sometime during this process the the cells within your body break down the mRNA into harmless pieces. **At no point does the mRNA in either vaccine change your DNA**. Getting an mRNA vaccine will not prevent you from being able to receive genetic therapy.

Always consult with your doctor for recommendations and if you have any questions regarding vaccines and COVID-19.

OTHER NORD INSIGHTS

A COVID-19 SURVEY OF THE COMMUNITY

Learn more at rarediseases.org/covid-19

772

Individuals with a rare disease, caregivers, and family members responded to the survey

98%

are worried about COVID-19

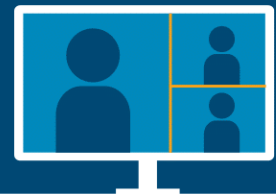


74%

Have had a medical appointment **CANCELED** due to COVID-19

59%

Have been offered a telephone or video call as an alternative to an in-office appointment



95%

of individuals and families have been impacted due to COVID-19



69%

Concerned about medication and medical supply shortages

29%

Have lost jobs temporarily or permanently due to COVID-19



11%

of job losses have resulted in loss of health insurance

NORD: Fighting for the rare community every day for more than 37 years.

NORD is committed to the identification, treatment and cure of rare disorders through programs of education, advocacy, research and patient support services. NORD does not recommend or endorse any particular medical treatment but encourages patients to seek the advice of their clinicians. NORD Headquarters: 55 Kenosia Avenue, Danbury, CT 06810 Tel: 203.744.0100 Fax: 203.263.9938 ©2020 NORD. All rights reserved. NORD® and RareInsights® are registered trademarks of The National Organization for Rare Disorders. NORD is a 501(c)(3) charity organization. NRD-2043

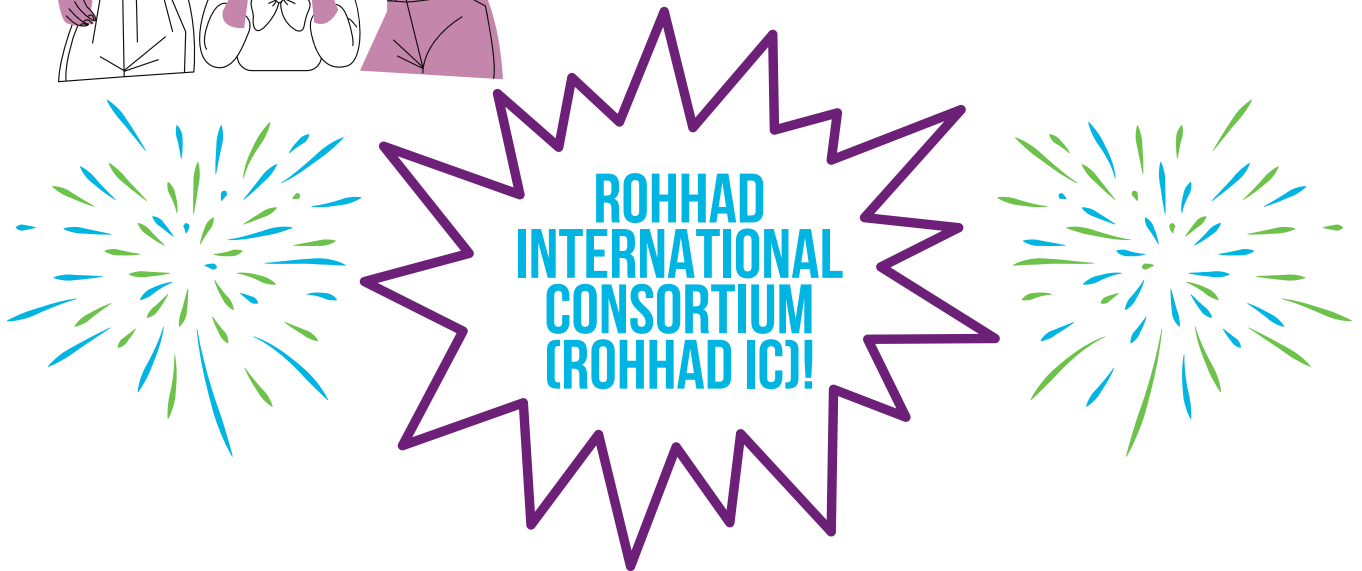


NORD[®]
National Organization
for Rare Disorders

rarediseases.org



In case you may have missed it, members of the ROHHAD community have been hard at work gathering a group of experts to develop the



"Our mission is to work strategically and collaboratively as clinicians, researchers, charities, and families by raising awareness, expediting diagnosis, optimizing care, and ultimately finding a cure for ROHHAD, a rare orphan syndrome."

ROHHAD community advocates like the ROHHAD Association, ROHHAD Fight Inc., the ROHHAD Association-Belgium, clinicians, and researchers saw a need to collaborate more effectively. Everyone stepped up to the challenge to establish this group.

We're excited to see all the great things ROHHAD IC will accomplish!

Learn more about by clicking the links below:
<https://www.rohhadfight.org/news>

[Dream team of scientists join forces to find ROHHAD disease cure thanks to Alexandria boy and his charity](#)



Q&A WITH A ROHHAD IC MEMBER: DR. MOHAMAD MAGHNIÉ

📍 GENOVA, ITALY

WHAT DOES YOUR DAY-TO-DAY SCHEDULE LOOK LIKE?

It is a struggle between what I want to do and what I have to do, trying to adjust my routine in balancing my time between administrative tasks, clinical activities and research.

The day is spent trying to answer a series of emails left behind, discuss clinical cases of patients with complex endocrine diseases and management meetings. My weekends are often dedicated to research.

WHY DID YOU BECOME A RESEARCH SCIENTIST?

My interest and curiosity for clinical science started early. To my view, It is a way of life rather than a profession where you grow daily by questioning and constantly learning. It is a lifelong personal desire for intellectual challenge.

WHAT LED YOU TO START STUDYING ROHHAD?

I became interested in ROHHAD after I met, as a third opinion, the first affected girl and the desperate mother reporting the changes she was seeing in her child.

According to the opinion of the other specialists, the child had simple obesity and the mother was neurotic.

WHY IS YOUR RESEARCH IMPORTANT?

It is a devastating disease that disrupts the lives of families. The diagnosis is late, the etiology is unknown, its evolution is multi-systemic and unpredictable, and therapy is only supportive. It is urgent to understand the pathophysiology to find answers and solutions that can save these children.



Q&A WITH A ROHHAD IC MEMBER: DR. JULIE HARVENGT

 LIÈGE, BELGIUM

WHAT DOES YOUR DAY-TO-DAY SCHEDULE LOOK LIKE?

I start my day by sharing a large healthy breakfast with my family. . . I mainly follow patients with rare genetic diseases . . . but I also look after patients in all areas of pediatrics. I am able to dedicate some days of the week to the development . . . in my research project to . . . study the epigenetic aspects of ROHHAD.

Also, as a young mother, I have to be on time to pick up my children after their school day!

WHY DID YOU BECOME A RESEARCH SCIENTIST?

Through my research activities I want to both impact the way I follow my own patients as well as to contribute to the growing body of research on this understudied condition.

Furthermore, studying the ROHHAD Syndrome is a rich and highly exciting field of research which may help us to understand new concepts; Not only for ROHHAD patients, but also for patients with other central dysregulations.

WHAT LED YOU TO START STUDYING ROHHAD?

I previously worked on a project oriented towards . . . childhood obesity, during which I met a patient suffering from ROHHAD.

I was confronted by the difficulties encountered by this patient and their family to establish the diagnosis and then the challenges we faced in managing the condition. Based on this experience I decided to focus my research activities on ROHHAD.

WHY IS YOUR RESEARCH IMPORTANT?

Our research is oriented to better understand which mechanisms are involved in the disease process among ROHHAD patients.

We hope that by gaining a better understanding of the mechanisms could lead us towards the development of new therapeutic approaches.

ROHHAD ORGANIZATIONS



ROHHAD SYNDROME JAPAN SECRETARIAT

HOME: SAITAMA PREFECTURE, JAPAN
FOUNDERS: MEGUMI HASHIMOTO AND HER SON KOUYA WHO WAS DIAGNOSED WITH ROHHAD AT THE AGE OF 4.

MISSION:

- RAISE RARE DISEASE AWARENESS IN JAPAN
- ESTABLISH A RESEARCH TEAM AND COLLABORATE WITH INVESTIGATORS OVERSEAS
- BRING TOGETHER AND EDUCATE JAPANESE FAMILIES AND PHYSICIANS

MORE INFO @
[THE ROHHAD SYNDROME JAPAN SECRETARIAT WEBSITE](http://www.rohhad.jp)



ROHHAD ASSOCIATION- BELGIUM

HOME: NEUPRE, BELGIUM
FOUNDERS: KIM BLYTH AND RUDY POLESE AND THEIR SON EDWIN WHO WAS DIAGNOSED WITH ROHHAD AT THE AGE OF 3.

MISSION:

- MAKE ROHHAD MORE KNOWN TO THE PUBLIC AND PHYSICIANS
- FINANCIALLY SUPPORT MEDICAL RESEARCH
- BRING TOGETHER BELGIAN AND INTERNATIONAL AFFECTED FAMILIES



MORE INFO @ [WWW.ROHHAD.BE](http://www.rohhad.be)

ROHHAD ORGANIZATIONS

ROHHAD FIGHT INC.

HOME: NEW YORK, USA

FOUNDERS: DANIELLE AND BILL CARNEY AND THEIR DAUGHTER MARISA WHO WAS DIAGNOSED WITH ROHHAD AT THE AGE OF 4.

MISSION:

- RAISE AWARENESS OF ROHHAD
- RAISE FUNDING FOR RESEARCH AND ROHHAD FAMILIES' TRAVEL EXPENSES AND MEDICAL COSTS

MORE INFO @ [HTTP://ROHHADFIGHT.ORG/](http://rohhadfight.org/)



ROHHAD ASSOCIATION

HOME: ALEXANDRIA, SCOTLAND

FOUNDERS: ELISABETH AND IAN HUNTER AND THEIR SON AARON WHO WAS DIAGNOSED WITH ROHHAD AT THE AGE OF 5.

MISSION:

- FUND AND PROMOTE ROHHAD RESEARCH
- OFFER SUPPORT FOR PATIENTS AND AFFECTED FAMILIES
- CAMPAIGN AND RAISE PUBLIC AWARENESS

MORE INFO @ [WWW.ROHHADASSOCIATION.COM](http://www.rohhadassociation.com)



GET INVOLVED!

AMAZONSMILE

The same great prices as regular Amazon, but with an amazing twist. Amazon will donate to a charity of your choosing every time you shop at AmazonSmile!

Shopping in the USA, Canada, etc. ?

Choose [ROHHAD Fight Inc.](#) as your charity to benefit.

Shopping in Europe or the UK?

Choose the [ROHHAD Association](#) as your charity to benefit.

CHARITY MERCHANDISE

Show your support for the ROHHAD Association!

Take a look at what's in stock [here!](#)

To place an order email: ROHHADAssociation@gmail.com

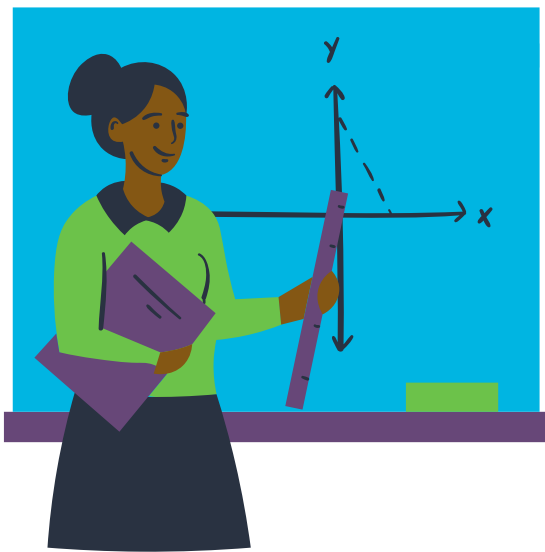
ZERO TO HERO: BECOME A ROHHAD AVENGER

1. REGISTER FOR ZERO TO HERO CHALLENGE
2. COMPLETE A CHOSEN DISTANCE FOR A SPORTING ACTIVITY IN 31 DAYS.
3. BECOME A ROHHAD AVENGER!

FOR MORE INFORMATION, VISIT THE ROHHAD ASSOCIATION WEBSITE [HERE.](#)

THANK YOU!

A *huge* thank you to all of the families, medical professionals, and scientists who have helped make this newsletter possible. We couldn't have created the ROHHAD Reader without you!



THIS ISSUE OF THE ROHHAD READER IS DEDICATED TO ALL OF THE FRONTLINE WORKERS ACROSS THE WORLD, WORKING EVERY DAY TO HELP US ALL IN THE FIGHT AGAINST THE DEADLY COVID-19 VIRUS