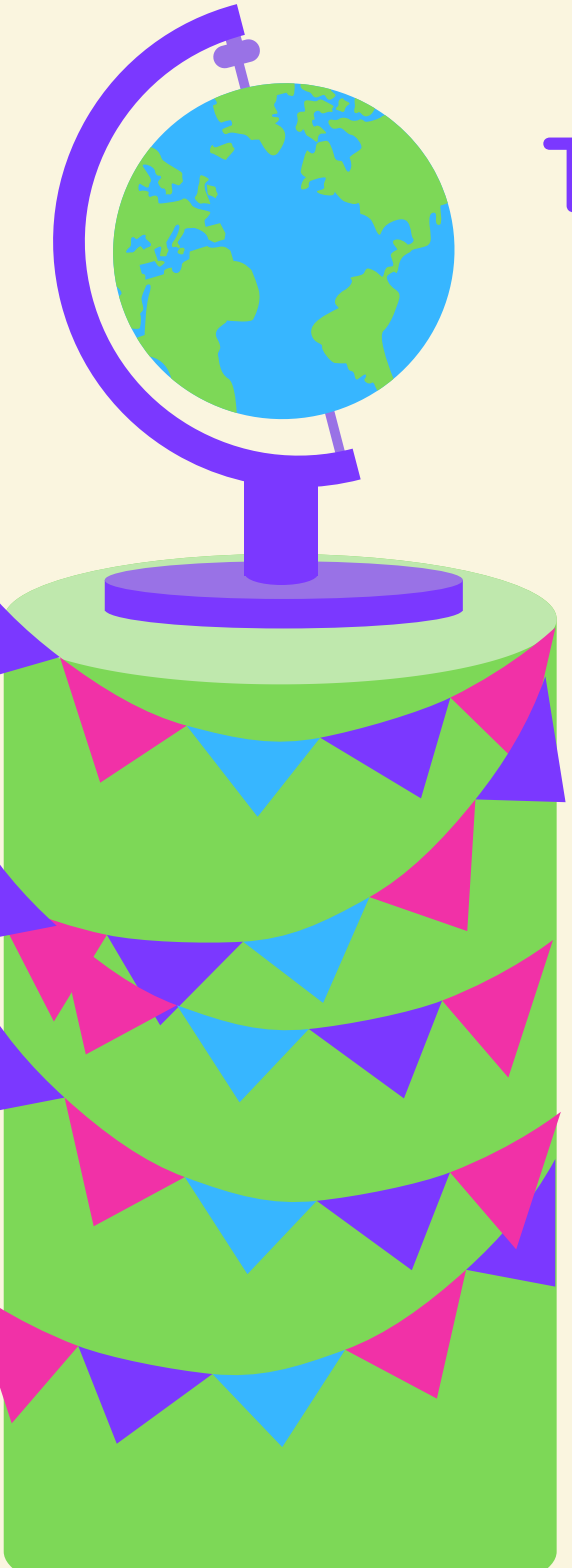
FEBRUARY 2020 | EDITION 4

# THE ROHHAD READER

RARE DISEASE DAY EDITION



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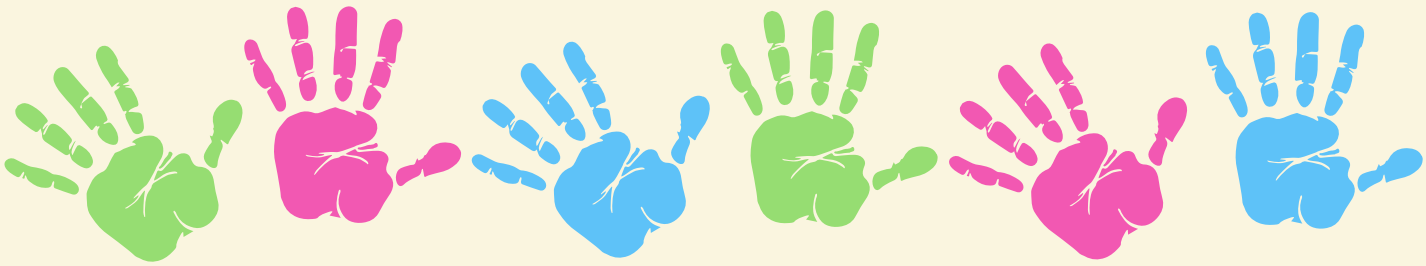
RAREDISEASEDAY.ORG

#RareDiseaseDay

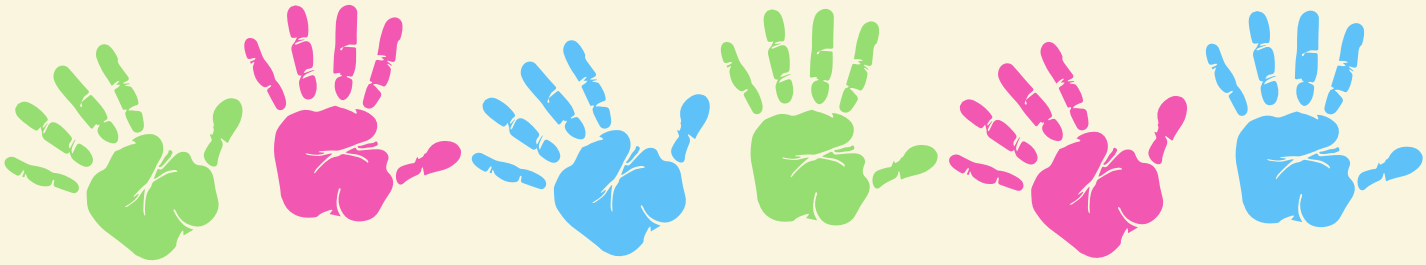


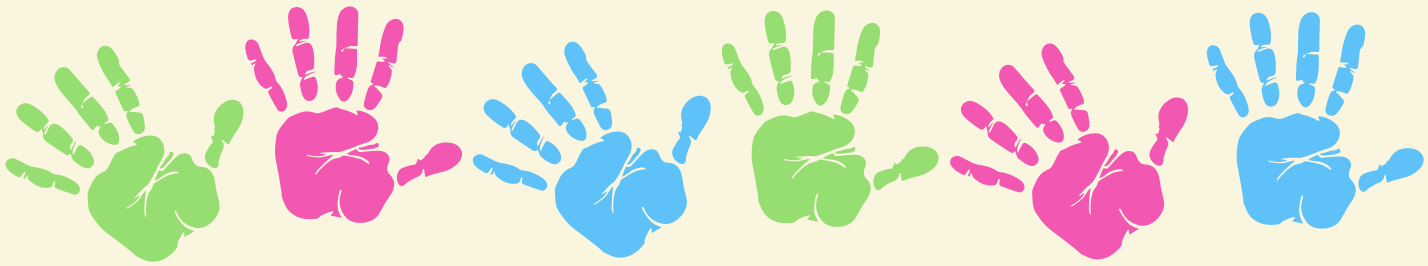
# welcome back!

We proudly present our fourth issue of the **ROHHAD Reader, a newsletter created for families whose daily lives are affected by ROHHAD.** Our goal is to teach and strengthen the ROHHAD community by bringing patients, families, physicians, and researchers together. This is a very special issue, our annual **Rare Disease Day** edition!

**February 29, 2020** will be the 13th International Rare Disease Day. This year, there will be **113 events in over 100 countries**!

#WeAreROHHAD



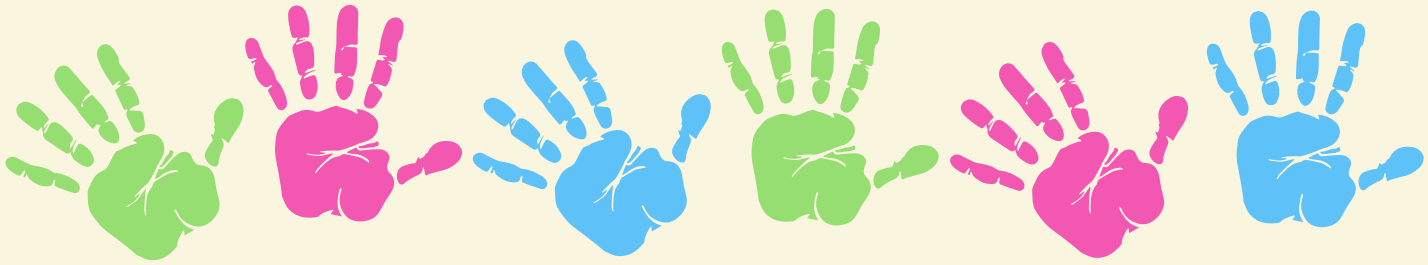


The goal of Rare Disease Day is to help provide equal access to diagnosis, treatment, and medical care for everyone living with a rare disease by raising awareness.

There are many ways you can help raise awareness! Attend an event near you. Follow #RareDiseaseDay and @rarediseaseday on Facebook, Instagram, and Twitter. You can also share your story on social media or on the official Rare Disease Day website.

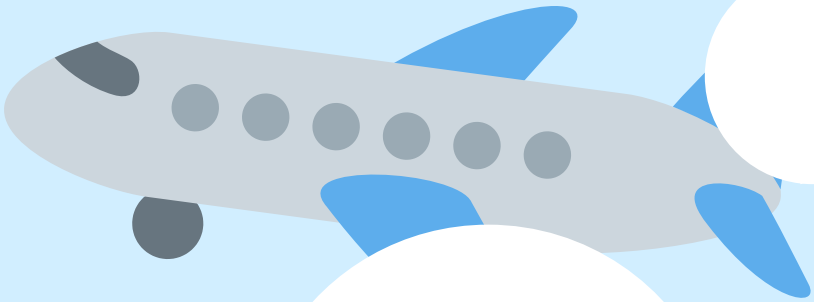
However you celebrate your day, we hope you get the opportunity to share your story with someone and that you get to learn something new about a rare disease!

Happy Rare Disease Day from,
*The ROHHAD Reader Team*

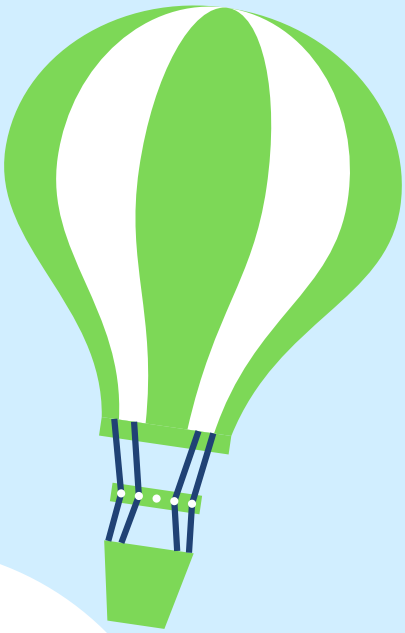


# Global Rare Disease Day Facts

There are over **6,000** rare diseases that over **300 million people**, from all over the world, live with.



**72%** of rare diseases are genetic. This means that they are not something you catch from someone else, like a cold, but may be something you were born with.



The first rare disease day was celebrated on **February 29th, 2008**. February 29th is a 'rare' date that happens only once **every 4 years**.

# Rare Disease Research

When you have a question, what do you do? Do you look up information online? What do you do if you can not find any answers?

When it comes to **rare diseases** like ROHHAD, doctors do not always have enough information to answer every patient's questions. This is when **medical research** can help!

Patients and their families are asked to participate in a research study and if they say yes, they become **participants**. Participants help by giving a researcher information about themselves or **data**.

A researcher might ask participants to answer questions in a **survey** or to give a small **sample** of blood or a baby tooth.

All these data help us find more answers to our questions!

# Success in Rare Disease Research

**Cystic Fibrosis (CF)** is a rare disease that causes a thick build-up of mucus in the organs. In the lungs, the mucus clogs the airways, causing breathing problems making it easy for bacteria to grow, often leading to early death.

In late 2019, a **breakthrough drug to treat CF was approved** for use in 90% of individuals with CF. This drug dramatically improves the quality of life for patients with CF. Doctors are hopeful this drug will offer these patients a **normal life expectancy.** *"Patients who were unsure about whether they should bother attending college because they had always known they would die young are now being told they should think about planning for retirement."*

**Where did this miraculous treatment come from?**
**Research!**

The new drug is the product of decades of research and advances, made possible through collaboration among academic researchers, patients, patient advocacy groups, and pharmaceutical companies.

Genetic studies identified that CF is caused by a mutation in a gene called CFTR. This discovery helped drive innovation leading to this new therapy. The breakthrough drug, called Trikafta, improves function of the defective CFTR gene.

**The results have been life-saving and life-changing!**

# ROHHAD Tooth Fairy Project

ROHHAD likely affects cells from the autonomic nervous system and the hypothalamus. These cells have been almost impossible to obtain for research. It was recently discovered that nervous system cells can be obtained from teeth, and these cells can be differentiated into hypothalamic neurons. By using cells from teeth, our research team wants to investigate changes in the way nervous system and hypothalamic cells from patients with ROHHAD function compared to those from people without ROHHAD.

## How can **YOU** help?

If your child with ROHHAD is starting to lose baby teeth or if you will be having teeth extracted in the near future, please email us at **CAMP@LurieChildrens.org** or call 312.227.6905 ***before*** the teeth have fallen out or been removed. This will allow us to mail a tooth collection kit to your preferred address ahead of time.

# Help the Tooth Fairy Find the Tooth!

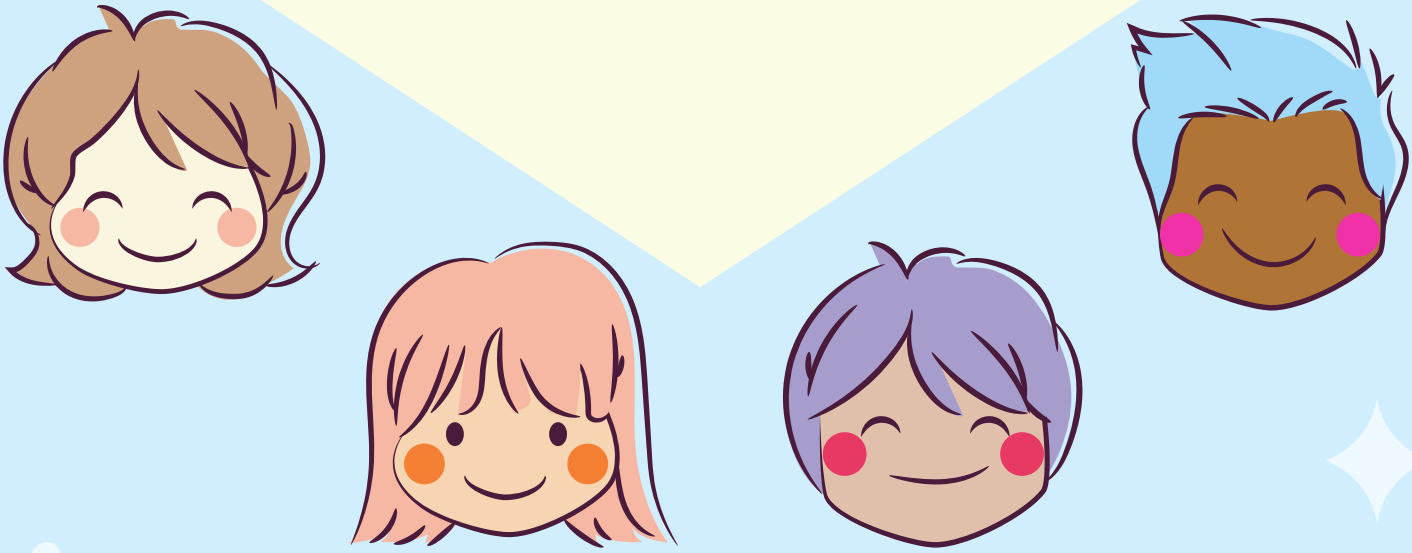
# Interested in getting more involved in ROHHAD research?

Join the ROHHAD Reader Community Advisory Board (CAB)! A CAB can help make a research project better by:

- Giving researchers advice on what research questions are the most important to ROHHAD families, and how we might answer those together
- Creating a space where participants can talk about their experiences with ROHHAD, to other patients and to the researchers
- Helping raise awareness for ROHHAD

*ROHHAD patients, caretakers or family members — all are welcome!*

Learn more by emailing us at CAMP@LurieChildrens.org

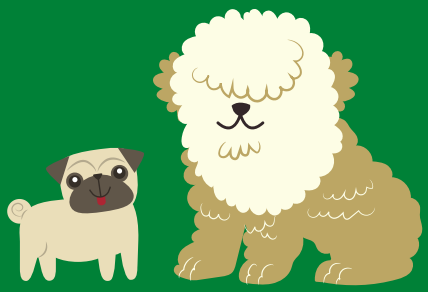


# ROHHAD Research

## Integrated Whole Genome and Transcriptome Sequencing

### What is it?

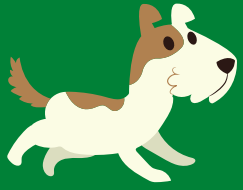
Researchers are looking to identify changes in the hypothalamus, a part of the brain responsible for controlling many autonomic nervous system functions like body temperature.



The team will collect blood cells from ROHHAD patients and their parents and turn them into hypothalamic neurons. Neurons are a type of cell found throughout the nervous system.

### How to help!

Contact Dr. Vidhu Thaker at **vidhu.thaker@columbia.edu** and you will be sent kits for blood collection.



# ROHHAD Research

## International NIH.gov ROHHAD Registry

### What is it?

The Registry is a secure database that contains information regarding the medical history and current health of patients with ROHHAD.

Participants will be asked to update or confirm their information at least twice a year.

### How to help!

To join the Registry, please click **here**. If you have not filled out the Registry within the past 12 months, please email **CAMP@LurieChildrens.org** and we will send you a secure link to update you or your child's current record.

# ROHHAD Research

## Exploring the Cause of ROHHAD Through Brain Inflammation and Hormones

### What is it?

There are reports of some ROHHAD patients with nervous system inflammation and some patients who respond to treatments that suppress the immune system, but the patients still go on to develop ROHHAD.

Normally, the immune system fights off invaders like viruses and bacteria. Sometimes the immune system can get confused and attack one's own body.

The team is studying whether an autoimmune attack on the nervous system may be the cause for some cases of ROHHAD.

### How to help!

Want more information? Please email **leslie.benson@childrens.harvard.edu** and **neurocore@childrens.harvard.edu** with "*BCC ROHHAD Research*" in the subject line.

# ROHHAD Research

## Tissue Banking

### What is it?

The Harvard Brain Tissue Resource Center (HBTRC), in collaboration with ROHHAD Fight Inc. and Lurie Children's Hospital, is coordinating registration of ROHHAD families for prospective post-mortem brain donations.

A post-mortem brain donation is a gift of knowledge that is of critical importance in furthering our understanding of ROHHAD and in helping future ROHHAD patients and families.

### How to help!

Read more about the tissue donation **here** and consider registration today.

# Rare Disease Day Events

EURORDIS Black Pearl Awards
Host: EUORDIS-RARE DISEASES EUROPE

Location: Le Plaza, Brussels, Belgium
Date: February 18th, 2020

An annual event that celebrates and recognizes those living with rare diseases as well as those who dedicate their time to making a difference in the rare disease community. Learn more about this event here!

Rare Disease Day Senedd Reception
Host: EUORDIS-RARE DISEASES EUROPE

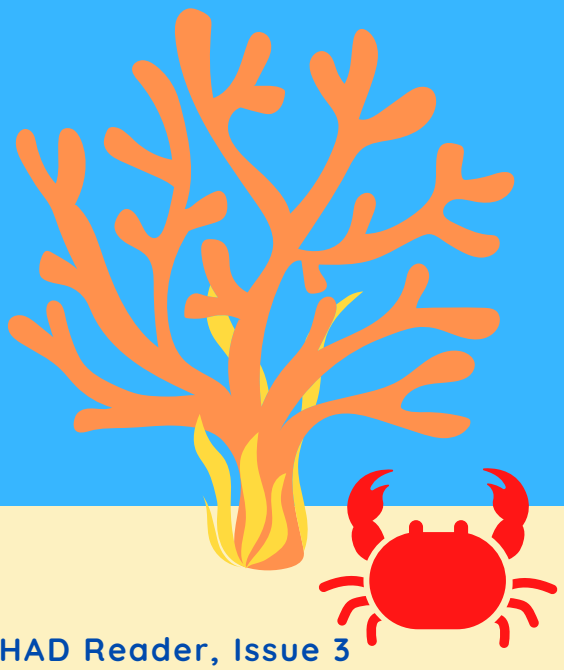
Location: Senedd, Cardiff Bay, UK
Date: February 25th, 2020

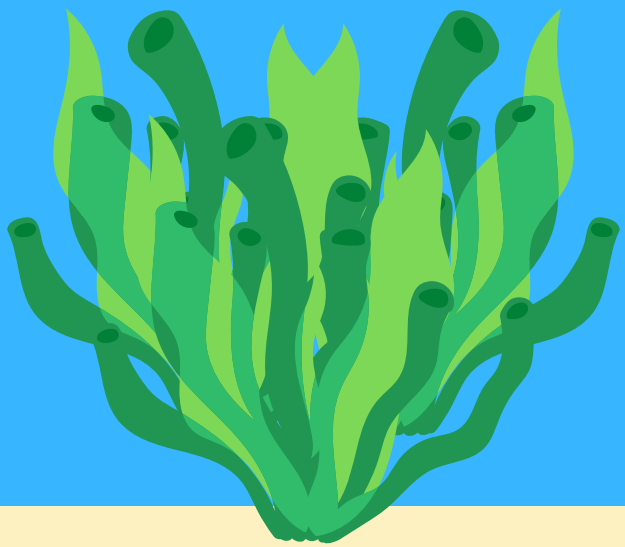
This event features several inspirational speakers to talk about important issues in the rare disease community, including an update from the Chair of the Rare Disease Implementation Group, Dr. Graham Shortland. Canapes and light refreshments will be provided. Register for this event here!

Rare Disease Day Westminster Reception
Host: Rare Disease UK

Location: London, UK
Date: February 26th, 2020

Rare Disease UK is the national campaign for people with rare diseases and all who support them. Speakers include: Baroness Blackwood, Dr. Jayne Spink, and Amanda Brodie. For more information, you can contact Genetic Alliance UK's Policy and Public Affairs Officer, Sophie Peet at sophie.peet@geneticalliance.org.uk.





# Rare Disease Day Events

Mission: Possible
Host: The National Brain Appeal

Location: London, UK
Date: February 29, 2020

A special evening of dinner and dancing to raise funds for Rare Dementia support. This event features a drink reception, three course dinner, entertainment and speeches, silent and live auctions, and dancing. Tickets are currently sold out, but you can join the waiting list here!

Red, Scarlet, Crimson: The History of Colour in Art
Host: Advocacy for Neuroacanthocytosis Patients

Location: London, UK
Date: February 29, 2020

This event features a tour of the V&A Museum to raise awareness of Rare Disease Day and Neuroacanthocytosis a group of rare disorders characterized by progressive muscle weakness and atrophy, etc. The tour explores the use of the color red in various artistic forms such as fabrics, jewels, and stained glass throughout European history. To make a reservation, contact annete@naadvocacy.org.

Joint North/South Rare Disease Day Conference
Host: The National Brain Appeal

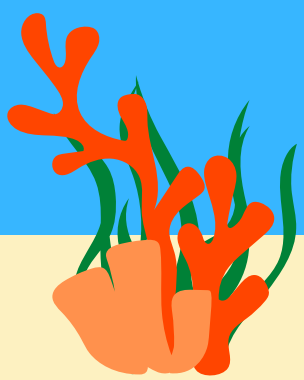
Location: Belfast, UK
Date: February 28, 2020

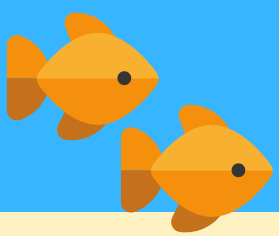
This event is for those who want to learn about the progress that has been made, what is currently going on, and next steps in the Rare Disease Day community. Register for this event here!

Cambridge Rare Disease Network's Rare-I-Tea Party
Host: Cambridge Rare Disease Network (CRDN)

Location: Cambridge, UK
Date: February 29, 2020

Cambridge Rare Disease Network (CRDN) invites you to join us for tea and cakes in celebration of this year's extra rare Rare Disease Day which falls in a leap year. We're delighted to be holding this friends and families event at the wonderful premises of the Cambridge charity Rowan Humberstone which supports adults with disabilities through art. Get free tickets here!





# Rare Disease Day Events

LA Clippers Game for Rare Disease Day
Host: California Rare Action Network

Location: Los Angeles, USA
Date: March 1, 2020

NORD and California Rare Action Network (RAN) will be featured as the Charity of the Night at the Clippers vs. Sixers game. California RAN will bring children affected by rare diseases on the court to meet NBA players. If you would like to attend the game and take part in this amazing Rare Disease Day event, tickets can be purchased here.

Rare Disease Research at Yale
Host: Yale University

Location: New Haven, USA
Date: February 26, 2020

At this event, you'll be able to learn about the rare disease research being conducted at Yale University. You'll get the chance to hear about inspirational patient stories and even meet the scientists involved in this great cause.

Topgolf Tournament for Myotonic
Host: Topgolf

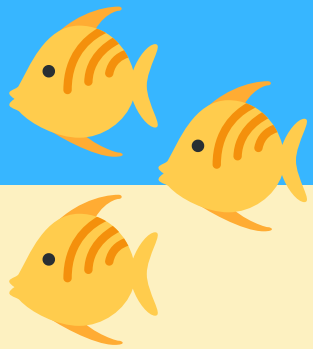
Location: Columbus, USA
Date: March 1, 2020

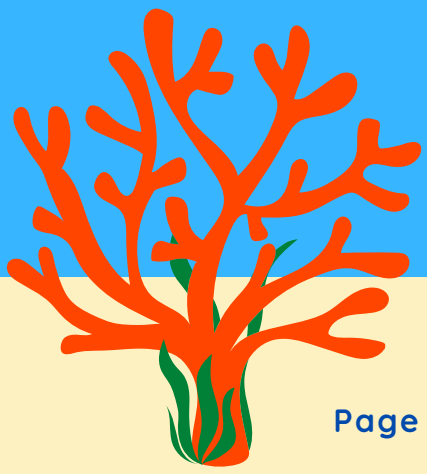
The event promises to be an exciting and entertaining day of great impact. Myotonic along with Paul & Sara Dillon's family and friends are organizing the event to raise awareness and much needed funding to support Myotonic's mission to enhance the quality of life of people living with myotonic dystrophy and advance research focused on treatments and a cure.

Rare Disease Day Symposium 2020
Host: Calvin University

Location: Grand Rapids, USA
Date: March 1, 2020

Come to this event to connect with others in the West Michigan community who are involved with or impacted by rare disorders. The goal is to bring all members of the rare disease community together ranging from patients, caregivers, families, researchers, advocates, and students. Find more information here!





# ROHHAD ORGANIZATIONS

## 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


## ROHHAD ASSOCIATION BELGIUM

**Home:** Neupre, Belgium

**Founders:** Kim Blyth, 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

## ROHHAD FIGHT INC.

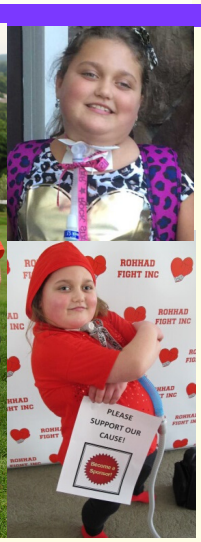
**Home:** New York, USA

**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/



# GET INVOLVED

## AmazonSmile

The same great prices as regular Amazon, but with an amazing twist. Amazon will donate to ROHHAD Fight Inc. every time you shop at AmazonSmile

1. Shop at AmazonSmile
2. Select ROHHAD Fight Inc.

NEW!

## Charity Merchandise

Buy charity merchandise in support of the ROHHAD Association

Email ROHHADAssociation@gmail.com to place an order

More information is available here.

## 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.

# Resources

ClinicalTrials.gov
www.clinicaltrials.gov

National Organization for Rare Diseases
rarediseases.org

ROHHAD Fight, Inc.
www.rohhadfight.org

ROHHAD Association
www.rohhadassociation.com

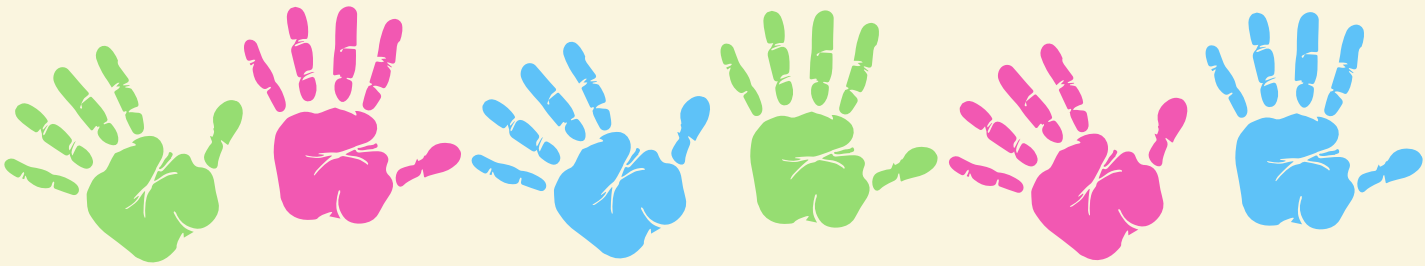
ROHHAD Association Belgium
https://www.rohhad.be

For More Information on
Cystic Fibrosis (CF) Research

Cystic Fibrosis Drug

The FDA Approves a
New Therapy for CF!

CF Foundation Celebrates
FDA Approval for Trikafta™



# thank you!

Thank you so much for taking the time to learn more about Rare Disease Day! We hope you enjoyed reading this edition as much as we enjoyed designing it!

We would like to give a huge thank you to our Clinical Research Coordinator, Valeria Islas Montantes, for all her efforts in putting together such a beautiful reader.

# for next time...

We need your help to make the next edition even more special! If you have any suggestions for improvement or if you have any stories, artwork, photos that you want us to feature in the next ROHHAD Reader, please send us an email at ROHHADReader@gmail.com.