



The ROHHAD Reader
EDITION 3
OCTOBER 2019

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Click [here](#) to check out our new Twitter page and follow #WeAreROHHAD for updates and announcements!

ROHHAD READERS
Welcome Back!



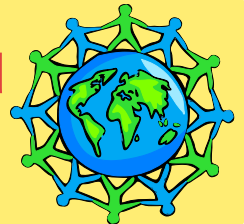
What is the ROHHAD Reader?



A biannual newsletter created for families whose daily lives are affected by ROHHAD.

Who's in it?

It features ROHHAD patients, families, and researchers from around the world.



Why was it created?



To inform and strengthen the ROHHAD community by bringing patients, families, physicians, and researchers together.

#WeAreROHHAD

ROHHAD FACTS



Weight gain of
20-30 lbs



in
3-6
months

Since 1965,
fewer than

150



cases of
ROHHAD have
been reported

40%
develop neural
crest tumors



Typical onset is
between ages of

1.5 to **10**
years



Half

require ventilator
support while asleep

.....

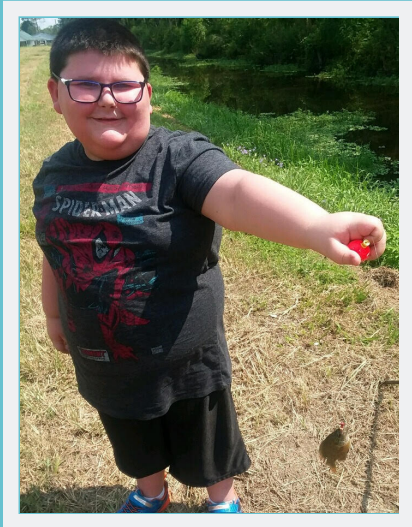
Half

require ventilator
support while asleep
and awake



Blaze

Age: 8



Blaze enjoys fishing with his Maw Maw Nita and Paw Paw Tim!

#WeAreROHHAD

FUN FACTS!



Aaron

Age: 9



Aaron is an avid collector. He started with transformers and Marvel and DC comics. Now he collects coins!



Sarah

Age: 27

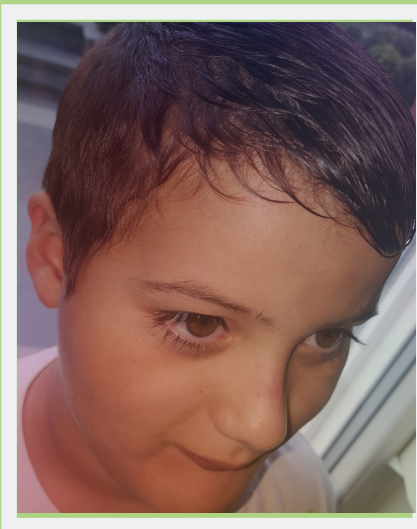


Sarah volunteers 30 hours or more a month with American Red Cross!



Edwin

Age: 7



Edwin dyed his hair purple like his favorite a cappella singer, Mitch from Pentatonix, who he hopes to meet!



Chance

Age: 15



Chance visited Family Guy studios and met Seth MacFarlane!

Announcing:

Rare Disease Day!

Saturday, February 29th, 2020.



What is it?

A day dedicated to raising awareness about rare diseases. Hundreds of rare disease organizations world-wide take part by hosting events, raising funds, and sharing information, all with the hope of advancing rare disease awareness and research.

When is it?

Every year, Rare Disease Day takes place on the **last** day of February, a month with a 'rare' number of days. Next year marks the 13th annual Rare Disease Day.

Save the date!

On **February 29th, 2020**, with the help of ROHHAD Fight, Inc and the ROHHAD Association, we will be hosting our very first online ROHHAD Rare Disease Day event. You won't want to miss out! Stay tuned for more information!

#RareDiseaseDay

#WeAreROHHAD

Did you know?

1 in 20 people live with a rare disease at some point in their life. You are not alone!

To learn more about Rare Disease Day click [here](#).

Patient Spotlight: **Blaze**

Age: 8

📍Hometown: Thibodaux, LA



Who do you admire?

"I admire my *Parrain Tim and my Uncle Chase. My Parrain Tim is a hard working dad with a very kind heart. My Uncle Chase is a firefighter and he is fighting cancer and he shows me that I can overcome my worst days with ROHHAD because he is fighting just like me."

*Parrain means godfather in French.

What is a goal you hope to accomplish in the future?

"...to play basketball with kids my age and one day I would like to become a firefighter like my Daddy and Uncle Chase"

What is the best advice you've ever received?

"Never give up & the sky is the limit."

What is an accomplishment you are proud of?

"I am very proud of myself for being able to get on the treadmill and stationary bike at therapy. When I first started I did not want anything to do with it because I was scared to fall. Now I get on it every Tuesday at therapy and I do 10 minutes on each."

Family Perspective

LeBouef Family

What is something you've learned from having a child with ROHHAD?

"We have learned that each day is different for him. He can go two days with his temperature normal and then we can go a few days with his temperature really low or really high. This syndrome has shown us how strong he really is. We have dealt with this syndrome for 5 years without knowing what was wrong with him. I feel that now that we know what is wrong with him I am able to educate myself and the community about what he goes through."



Patient Spotlight: **Chance**

Age: 15

📍 Hometown: Wickliffe, KY

What do you like to do in your free time?
"Watch Chrisfix videos on Youtube"



What is a goal you hope to accomplish in the future?
"Graduating high school"

Describe a happy memory.

"Visiting the Family Guy Studios and meeting Seth MacFarlane in person and having lunch with him"

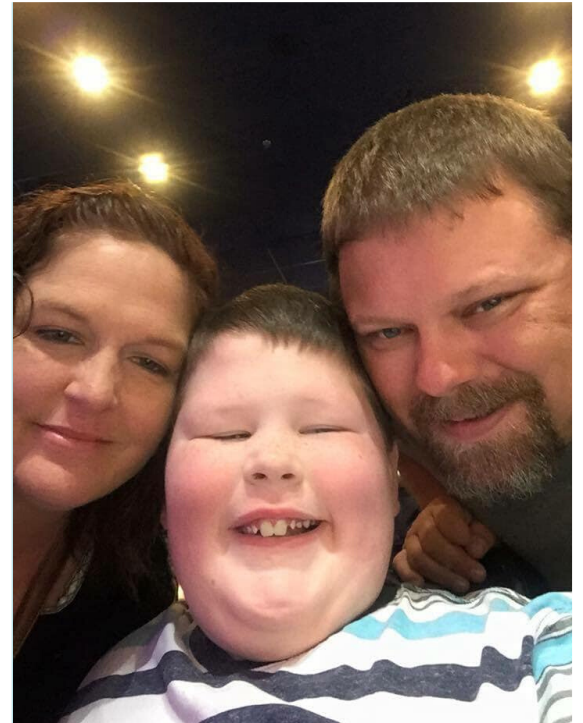
What do you enjoy learning about in school?
"Science and Math"

Family Perspective

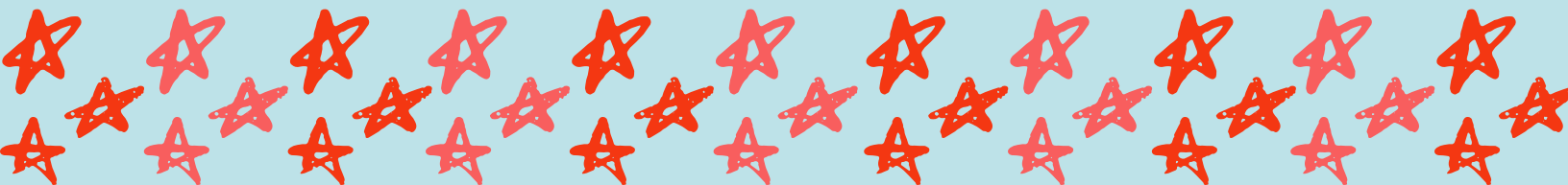
What is something you've learned from having a child with ROHHAD?

Yon
Family

"Never take even one second for granted, cherish the small things in life, stop and take the time to enjoy the rainbows, the butterflies, the flowers. Our milestones and accomplishments may not look like the typical families' but they are just as important and special. Always kiss them goodnight, say a prayer and thank God in the morning when you hear 'Hey, Mom!' "



If you are interested in being a patient spotlight in the next edition, please email rohhadreader@gmail.com



Patient Spotlight: Harper

Age: 6

📍 Hometown: Rathdrum, ID



Describe a happy memory.

"Going to the 'rock beach' with my family. It is my favorite beach to go swimming with my papa."

What is an accomplishment you are proud of?

"I worked hard practicing reading all summer. This year in school I tested at the very top of the scale for reading!"

What do you like to do in your free time?

"I like to dance and swim. I have been dancing for 4 years and swimming for 2!"

What are you most thankful for?

"I am thankful for my friends and family. I love to love and I love to give hugs. It makes me happy to make other people's hearts happy."

Family Perspective

Pursley Family

What was your diagnosis journey like?



"Shortly after Harper's 4th birthday we started noticing changes. She gained about 25 pounds in 4 months, was exhausted all the time, and had severe social anxiety. We contacted her doctor, and he listened to me and didn't play off her symptoms as a "growth spurt" or us over feeding her. We ran several tests, and what gave us our first real glimpse at our diagnosis was a sleep study that showed Harper's O2 levels dropping into the 50s.

The next day we were sent home with oxygen and instructed to use it whenever Harper slept. Two days later, I walked in her room to Harper making almost a choking noise. She was unresponsive and it took us 15 minutes to get her to open her eyes. We were then transported to our local ER where the pediatric specialist made the decision to life flight Harper to Seattle Children's hospital to get our formal diagnosis.

Our journey to a diagnosis was so much quicker than a lot of other families. What takes other families years to get, took us just over 2 months thanks to her amazing pediatrician and a mom's intuition."

Meet our Junior Editor!

Sarah Mineau

Age: 27

📍 Hometown: Marysville, MI



What is a challenge you have faced and what have you done to overcome it?

"A big challenge I have overcome and still deal with is people asking me when I am going to deliver or am due. With ROHHAD, I carry the weight in my stomach and being the age I am people usually just "assume" before they know the facts. Now that I have gotten older, I explain...what ROHHAD is."

What is a goal you hope to accomplish in the future?

"To write a story or get a blog going about my story with ROHHAD. I want to educate others and give those that are younger hope."

What is an accomplishment you are proud of?

"I have received 2 associate degrees from my local community college: one in Liberal Arts and the other in Business. "

What advice do you have for other people with ROHHAD?

"Don't give up the fight, there is a reason we have this. LISTEN to your body and anything weird, different or you are unsure about. Talk to a doctor."



Q&A with a Researcher

Dr. Mehul Dattani

 London, England

Why did you become a research scientist?

"I wanted to understand in greater depth the rare clinical conditions affecting my patients. I wanted to understand how they developed the conditions, and importantly, I wanted to develop novel therapies that would lead to an improved quality of life for them."

What does your day to day schedule look like?

"I start the day trying to grapple with more than 300 emails. I then do ward rounds and clinics with a number of management meetings. I look after patients with many unusual endocrine conditions... My research is left to the evenings and weekends..."

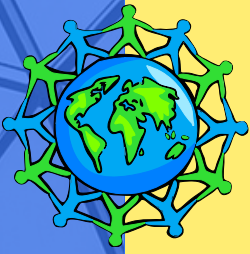
What led you to start studying ROHHAD?

"I was referred patients with this challenging condition. The resilience of my patients amazed me, and I was frustrated by the fact that I had to deal with the challenging symptoms that they manifested, yet did not understand why the condition occurred. I felt that research was urgently needed."

Why is your research important?

"ROHHAD is a devastating condition and we need to do all we can to improve the lives of these young patients... Additionally, we need to understand why the condition occurs before treatments...can be developed."





ROHHAD RESEARCH IS TAKING PLACE ALL AROUND THE WORLD!

CAN YOU MATCH THE RESEARCH SITES AND ORGANIZATIONS SUPPORTING THIS RESEARCH TO THEIR PINS?



EACH COLOR REPRESENTS A DIFFERENT COLLABORATION WHOSE RESEARCH IS DISCUSSED IN THIS ISSUE!



Chicago



London



New York



Boston



Calgary



Glasgow



San Francisco



Memphis



Genoa



Houston

EACH COLOR BELOW REPRESENTS A DIFFERENT ORGANIZATION RAISING FUNDS TO SUPPORT RESEARCH



ROHHAD Association



ROHHAD Fight Inc.



ROHHAD Association Belgium



The Tooth Fairy Project



Recent Discovery

ROHHAD affects cells from the nervous system. These cells have always been almost impossible to obtain for research. It was recently discovered that these cells can be obtained from lost teeth.

What's Next?

The team will investigate changes in the way nervous system cells from ROHHAD patients function compared to those from people without ROHHAD.

How can **YOU** help?

If your child with ROHHAD is losing baby teeth, or if they plan to have any teeth taken out, please email CAMP@luriechildrens.org and we will send you a tooth kit in advance.

International ROHHAD Registry

What is it?

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

What's Next?

Capturing information on as many patients as possible, including updated information from year to year will help us learn more about ROHHAD and guide research into treatments.

How can **YOU** help?

To join the registry, click [here](#). If you have previously filled out the registry, but not in the last year, please email CAMP@luriechildrens.org, and we will send you a secure personal link to update your current record.



Investigating Causes and Symptoms of ROHHAD

What is it?

The researchers have identified *genetic variants in ROHHAD patients that may play a role in ROHHAD, and they are collaborating to study these variants.

What's Next?

Using these variants, the researchers plan to create models of ROHHAD which they can use to test potential new therapies for ROHHAD. They will also study the immune system in patients with ROHHAD to see if it functions differently.

Additionally, they plan to collect information regarding the medical history of ROHHAD patients in order to examine relationships between the genetic variants and clinical phenotype.



What's a *genetic variant?

An alteration in one of your genes. While some variants are harmless, others can be linked to diseases.



Research Update



Integrated whole genome and transcriptome sequencing



What is it?

Researchers are looking to identify changes in the *hypothalamus that may be responsible for ROHHAD.

What's Next?

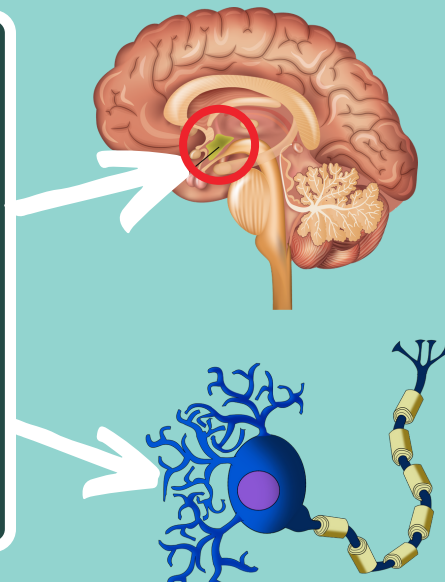
The team will collect blood cells from ROHHAD patients and their parents and turn them into hypothalamic *neurons. Then, they will look for genetic differences between the neurons.

How can YOU help?

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

***Hypothalamus:** A small region of the brain responsible for controlling many functions including body temperature, thirst, hunger, and hormones.

***Neuron:** A type of cell found throughout the nervous system. The nervous system includes your brain and spinal cord.



For more information on the study, please click [here](#).



Exploring the cause of ROHHAD through brain inflammation and hormones

What is it?

There are reports of ROHHAD patients with nervous system inflammation and response to treatments that suppress the immune system. Consequently, the team is studying whether an *autoimmune attack on the nervous system may be the cause for ROHHAD.

What's Next?

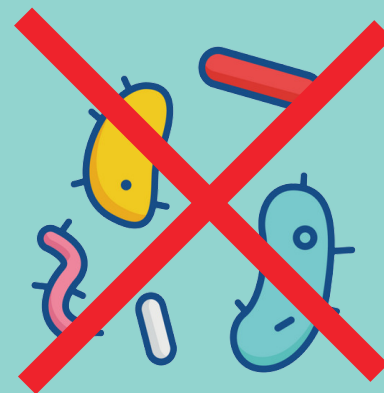
The team is interested in storing blood, spinal fluid and tumor tissue in order to explore what causes ROHHAD, how ROHHAD works, and ultimately develop better treatments.

How can **YOU** help?

To learn more about contributing samples to the research, please email leslie.benson@childrens.harvard.edu and neurocore@childrens.harvard.edu with "BCC ROHHAD Research" in the subject line.

What's an *autoimmune attack?

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



Research Update

Tissue Banking for ROHHAD Research

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 in helping future ROHHAD patients and families.

How can **YOU** help?

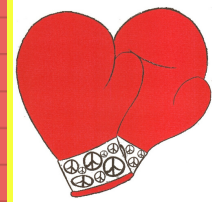
Although we know death is a difficult thing to consider, the time to start thinking about donation is now. Early discussion reduces the stress of such decisions at the time of death and allows for arrangements to be set in advance.

Read more about the bank [here](#) and consider registration today.





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 @<http://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 @<https://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](#).

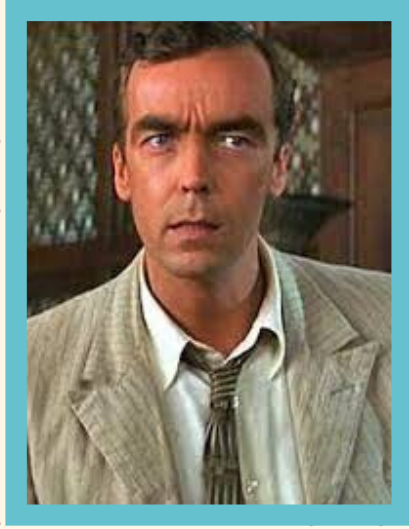
ZERO TO HERO

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](#).

COMMUNITY AWARENESS



The Mummy actor **John Hannah** supports **#ZeroToHeroChallenge**



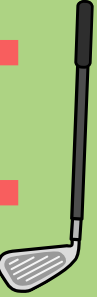
Robert Downey Jr. supports **#GoMadforROHHAD**



Recent Events

The 9th Annual Golf Tournament

Hosted By: ROHHAD Fight Inc.
July 6th, 2019



Glitter Ball

Hosted By: The ROHHAD Association
September 7th, 2019



Recent Events

Pride of Britain Awards

Partnered by ITV, The Daily Mirror and TSB

October 28, 2019



Aaron Hunter, from the ROHHAD Association, received the **Good Morning Britain Young Fundraiser of the Year** at the Pride of Britain Awards. This is an event that honors extraordinary people that are working towards making the world a better place. Aaron was recognized for helping raise an astonishing **£310,000 (\$400,000 USD)**! The money raised will be used to help researchers from all over the world find a cure for ROHHAD.

All of us at ROHHAD Reader would like to congratulate you Aaron!
Thank you for all that you have done to help out our ROHHAD community!

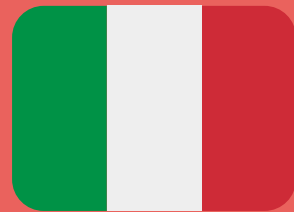
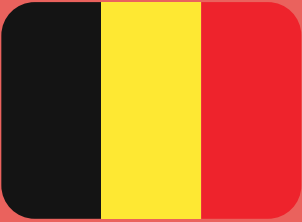
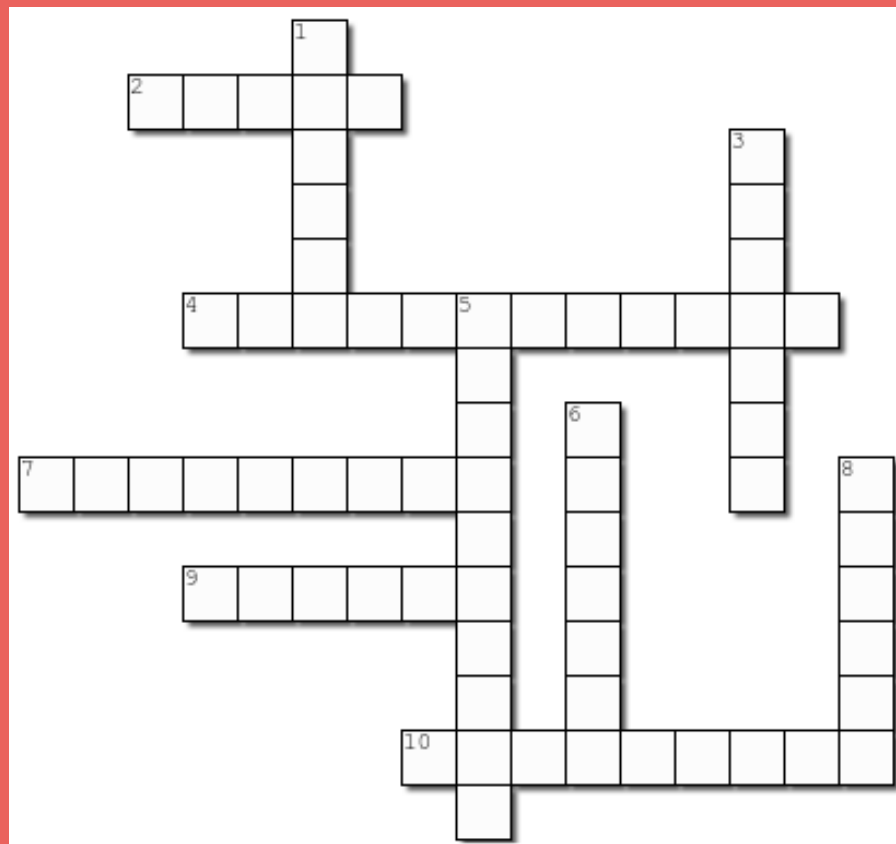


You're our superhero!



Crossword Puzzle

-ROHHAD Cities-



ACROSS

1. Researchers here are collaborating with researchers in London and Glasgow
3. Researchers here are collaborating with researchers in Boston
6. Hometown of Blaze
8. Hometown of Dr. Dattani
9. Hometown of Chase

DOWN

1. Researchers here are studying autoimmune attacks on the nervous system
3. Location of Ann & Robert H. Lurie Children's Hospital of Chicago
5. Where ROHHAD Association is located
6. Where ROHHAD Fight Inc. is located
8. Where ROHHAD Association Belgium is located

Answers: Across: 1. Genoa, 3. San Francisco, 6. Thibodaux, 8. London, 9. Wickliffe, Down: 1. Boston, 3. Chicago, 5. Alexandria, 6. New York, 8. Neupre

Answers:



Resources



ClinicalTrials.gov

<https://clinicaltrials.gov>

National Organization for Rare Diseases

<https://rarediseases.org>

ROHHAD Fight, Inc.

<https://www.rohhadfight.org>

ROHHAD Association

<https://www.rohhadassociation.com>

ROHHAD Association Belgium

<https://www.rohhad.be>

thank you!

A huge thank you to all of the families who contributed pictures, responses, and inspiring stories to this edition.

We couldn't have created this without you!

A special thank you to Danielle Carney and Elisabeth Hunter for reaching out to the ROHHAD community worldwide, and for their tireless efforts to continue to advance ROHHAD awareness.

Finally, a huge thank you to undergraduate students Grace Tu and Nikita Menta for their amazing contributions to the Reader.

To view the past two editions of the ROHHAD Reader, click the links below:

[Issue 1](#)

[Issue 2](#)

We would like to dedicate this issue of the ROHHAD Reader to Sedona, who passed away in 2015.

