

THE

ROHHAD READER

RARE DISEASE DAY EDITION

FEBRUARY 28, 2019



TABLE OF CONTENTS

What is Rare Disease Day?.....	3
What is ROHHAD?.....	4
Patient Spotlight.....	5
Recently in ROHHAD.....	7
Ongoing Research.....	9
Raising Funds and Awareness.....	10
Community Organizations.....	11
Resources.....	12
Thank you!.....	13

WELCOME BACK!



Dear ROHHAD Readers,

Welcome back! We are excited to present you with the second issue of the **ROHHAD Reader**, a biannual newsletter designed specifically for families whose everyday lives are affected by Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (**ROHHAD**).

This Thursday, February 28, 2019 is the twelfth annual **Rare Disease Day**. **Rare Disease Day** is an international campaign that encourages people everywhere to make some noise and raise awareness for all types of rare diseases (see pg. 3).

With only over 100 cases of **ROHHAD** identified worldwide, **ROHHAD** is a very rare disease. We hope that this issue of the **ROHHAD Reader** will help families across the world participate in **Rare Disease Day** to raise awareness for **ROHHAD** and the way that it affects people's everyday lives.

Happy Rare Disease Day from,
The ROHHAD Reader Team

WHAT IS RARE DISEASE DAY?



The very first **Rare Disease Day** was held on February 29, 2008. The founders of Rare Disease Day selected this date because February 29 is a "rare" date that only happens once every four years. Ever since the inaugural celebration, Rare Disease Day has been held on the last day of every February. In 2008, only a few European countries participated. Each year since then, though, more and more cities and countries have joined the campaign.

This year, **93** countries will host **430** Rare Disease Day events !

The purpose of these Rare Disease Day events, and other Rare Disease Day initiatives, is to raise awareness about rare diseases and the effect that they have on so many people worldwide.

The goal is to encourage neighbors and friends, policy makers and researchers to learn more about rare diseases and the ways they can help contribute to finding a cause, a cure, or even just a little bit of new information.

#SHOWYOURRARE
#SHOWYOU CARE

How you participate in Rare Disease Day is totally up to you!



LEARN MORE @

<https://www.rarediseaseday.org/>

WHAT IS ROHHAD?

Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (**ROHHAD**) is a rare disorder that affects the autonomic nervous and endocrine systems.

The **autonomic nervous system** is responsible for many of the things that our body controls automatically, like our heart rate, body temperature, respiratory rate, sweat production, and digestion.

The **endocrine system** is responsible for sending out signals called hormones that stimulate growth and regulate metabolism. In people with ROHHAD, neither the autonomic nervous system nor the endocrine system function normally.



Some potential symptoms of ROHHAD:

- Significant weight gain over a 6-month period in children between the ages of 1.5 and 10 yrs.
- Hypothyroidism (underactive thyroid gland)
- Early or late-onset puberty
- Inability to regulate body temperature or maintain a normal water balance
- Insufficient ventilation with shallow breathing during sleep

Please keep in mind that this list is not all-inclusive and includes many symptoms that can present differently in different children.

There is, still, no known cause or cure for ROHHAD. However, research teams around the world continue to collaborate on studies that aim to increase our understanding of the disorder and how to most effectively treat and care for patients who are affected by it (see pg. 9).

PATIENT SPOTLIGHT

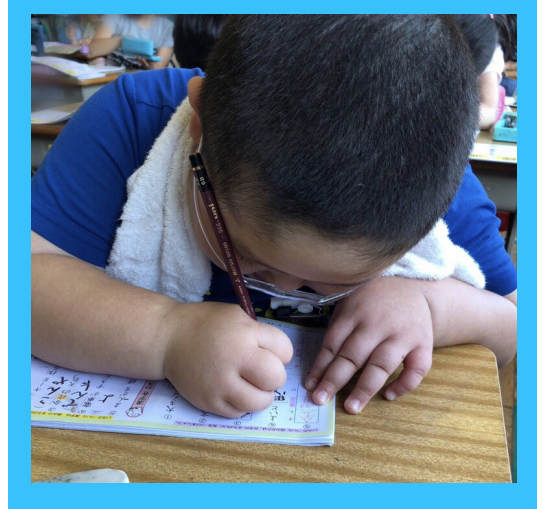
Meet **KOUYA**

Age: 8 yrs. Home: Japan

Tell us about your school.

"I go to elementary school on weekdays. I am trying especially hard to study **Kanji**. I am happy to see the smile of my classmates and I am having fun school life every day."

Kanji is a system of Japanese writing using Chinese characters.



家

HOUSE

児

CHILD

本

BOOK

玩

PLAY



Tell us about an activity you enjoy.

"I am learning **rugby** every Sunday. It's very fun. The dream of the future is a rugby player. The reason is because it is very cool."

Rugby is a team game played with an oval ball that may be kicked, carried, and passed from hand to hand. Points are scored by putting the ball on the ground behind the opponent's goal line or by kicking it between the two posts of the opponent's goal.

PATIENT SPOTLIGHT

From, **KOUYA's** Mom

"**Kouya** is in elementary school, not different from other children, moving physical in physical education, singing songs and playing musical instruments in music, and working hard on studying, but. . . Kouya lives with various risks. . . "

"Our family wants **Kouya** to live the same life as an ordinary child as much as possible. He wants it, too. And above all, in order to continue **Kouya's** favorite **rugby** for a long time in the future, based on the cooperation of the whole family, he seriously worked on a diet. Although it was said that it is impossible for ROHHAD patients, **Kouya** succeeded in losing 6 kg. [13.2 lbs.] in half a year."

"I would like to continue supporting **Kouya**, who will challenge in the future without giving up anything in the future. I am proud of **Kouya**. This **love** never changes."



ROHHAD + Anesthesia

In September 2018, a clinical team at the Ann & Robert H. Lurie Children's Hospital of Chicago in Illinois, USA published an article about how a group of their patients responded to **anesthesia** during surgery. One of the patients in this group had ROHHAD. The doctors who wrote the paper reviewed their patients' medical records and identified a number of potential challenges associated with using **anesthesia** in children with ROHHAD and other similar disorders. They concluded that while it is possible to use **anesthesia** safely in these children, it is also important that the doctors who administer the anesthesia are aware of the potential challenges.

[Read a summary of the article HERE.](#)

In November 2018, a clinical team at Ramón y Cajal University Hospital in Madrid, Spain published an article titled "Anesthesia in a Pediatric Patient with ROHHAD Syndrome." This article also showed that using **anesthesia** in children with ROHHAD requires special care, consideration and planning.

The doctors who wrote the article noticed that, after they administered anesthesia to their patient with ROHHAD, their patient experienced a decrease in his breathing rate, a decrease in the amount of oxygen in his blood, an increase in the amount of carbon dioxide in his blood and, eventually, episodes of **apnea**. These observations caused the doctors to change the type of anesthesia they were using in order to make sure their patient with ROHHAD was safe.

[Read a summary of the article HERE.](#)

Both of these articles were published in medical journals to help more doctors become aware of the fact that children with ROHHAD might respond differently to anesthesia than children without ROHHAD.

If you or your child is scheduled to receive anesthesia, it is a good idea to remind the doctors that you or your child has ROHHAD and may require special considerations.

Anesthesia is a kind of medicine that is administered before some medical procedures in order to make patients sleepy and insensitive to pain.

Apnea is a temporary pause in breathing.



A New ROHHAD Awareness Video

In February 2019, the **ROHHAD Association** collaborated with Robert Downey Jr., children with ROHHAD, and families around the world to create a brand new video to help raise awareness for ROHHAD.



You can share the video link below with your family and friends or post it on social media. Helping people learn more about ROHHAD is one great way to raise awareness!

<https://youtu.be/a-jXa5yzRNc>

Ongoing Research

We would like to extend a **BIG THANK YOU** to everyone who has contributed to any of the ongoing ROHHAD Research Studies.

Research teams in many different countries continue to search for better ways to diagnose and treat ROHHAD. Below are brief summaries of some ongoing ROHHAD Research Studies that need your help!

1. International ROHHAD Registry + Biomarkers

The goal of this project is to collect and study information about symptoms of ROHHAD and about biological samples, such as blood or saliva, from people with ROHHAD and their family members. Studying these two things together will help researchers understand how ROHHAD develops, how it affects people's bodies, and how it can be treated.

Read More @ <https://www.luriechildrens.org/en/specialties-conditions/autonomic-medicine/research/rohhad-studies/>

2. Metabolic Phenotyping in ROHHAD

This research seeks to understand how and why changes in energy balance within the bodies of people with ROHHAD might lead to the symptom of rapid-onset obesity.

Read More @ <https://clinicaltrials.gov/ct2/show/NCT02602769?cond=ROHHAD&rank=3>

3. ROHHAD Banking

This study was designed to collect post-mortem samples in order to empower future research to determine the cause of ROHHAD. These samples are critical to furthering an understanding of ROHHAD in order to help patients and their families in the future.

Read More @ <https://hbtrc.mclean.harvard.edu/>

4. ROHHAD European Consortium

This study plans to investigate the causes and symptoms of ROHHAD by studying genetics and physiology. The researchers working on this project will investigate new therapies and methods of diagnosis. **Look out for more information about how you can participate in this research soon!**

Raising Funds and Awareness

Research Funding

The **ROHHAD Association** has received many applications for funding for ROHHAD research. Look out for summaries of funded projects in the next issue of the ROHHAD Reader.

ROHHAD Fight Inc. Panera Bread Fundraiser New York, USA

When? February 28, 2019, 4:00pm - 8:00pm

Where? 5191 Sunrise Highway, Bohemia, NY 11716

How? Bring the flyer linked [here](#).



This March, dads, uncles, brothers, cousins, and friends of children with ROHHAD are waxing their legs, backs, arms, or chests to raise money and awareness.

Check it out on Facebook [HERE](#).

Email rohhadassociation@gmail.com to sign up.

AMAZON SMILE

AmazonSmile is an Amazon website with the same products, prices, and features as regular Amazon, but with one special addition. Every time you shop on AmazonSmile, you can request that Amazon donates a portion of the purchase price to **ROHHAD Fight, Inc.** or the **ROHHAD Association**

<https://smile.amazon.com/>

#SHOWYOURRARE

Participate in this year's **Rare Disease Day** social media campaign.

1. Paint your face
2. Pose for a selfie or group photo
3. Post the picture on social media using the hashtags **#ShowYourRare** and **#RareDiseaseDay**

Read more [HERE](#).

ZERO TO HERO Challenge

The inclusive sporting challenge that gives you control. You choose your sport.

You choose the distance. You choose a team or individual challenge. You choose the month. Read more and sign up on the

ROHHAD Association's website [HERE!](#)

Go Mad, Wax for ROHHAD

COMMUNITY ORGANIZATIONS

A few of the community organizations supporting ROHHAD

ROHHAD FIGHT INC.

Home: New York, USA

Founders: Danielle and Bill Carney, and their 12-year-old daughter, Marisa, who was diagnosed with ROHHAD when she was 4 years old

Mission: Expand knowledge about ROHHAD, advance treatment, discover a cure, enhance the quality of life of children with ROHHAD and ease the financial burden of families who have children suffering from ROHHAD

CHECK THEM OUT @ <http://rohhadfight.org/>

ROHHAD ASSOCIATION

Home: Alexandria, Scotland

Founders: Elisabeth and Ian Hunter and their 8-year-old son, Aaron, who was diagnosed with ROHHAD when he was 5 years old

Mission: Fund and promote research, help and support families, and campaign and raise awareness for ROHHAD

CHECK THEM OUT @ <http://www.rohhadassociation.com/>

ROHHAD ASSOCIATION BELGIUM

Home: Neupré, Belgium

Founders: Kim Blyth, Rudy Polese, and their 6-year-old son, Edwin, who was diagnosed with ROHHAD when he was 3 years old

Mission: Publicize ROHHAD to encourage early detection and increase survival, and fundraise for research to find a cause and improve treatment

CHECK THEM OUT @ <https://www.rohhad.be/>

If you know of any additional community organizations that might like to be featured, let us know!

RESOURCES

Rare Disease Day

<https://www.rarediseaseday.org/>

ClinicalTrials.gov

<https://clinicaltrials.gov/>

National Organization for Rare Diseases

<https://rarediseases.org/>

ROHHAD Association

<http://www.rohhadassociation.com/>

ROHHAD Association Belgium

<https://www.rohhad.be/>

ROHHAD Fight Inc

<http://rohhadfight.org/>

THANK YOU!

Again, the ROHHAD Reader Team says **THANK YOU!** Thank you to all of the children and adults with ROHHAD, families, friends, researchers, and physicians who contributed to this **Rare Disease Day** Edition of the **ROHHAD Reader**.

NEXT TIME...

We need your help creating the next edition of the **ROHHAD Reader!** If you have any stories or poems, artwork or photographs that you would like to have featured in the next **ROHHAD Reader**, or if you are interested in helping to write, design, or create the next issue, send us an email at **ROHHADReader@gmail.com**.

