OCTOBER 2018 ISSUE 1

THE ROHHAD READER

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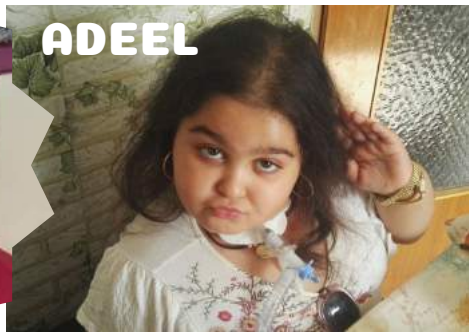


WHAT IS THE ROHHAD READER?

Dear ROHHAD Readers,

Welcome! This summer, the ROHHAD Association, ROHHAD Fight Inc., and the Center for Autonomic Medicine in Pediatrics (CAMP) at the Ann & Robert H. Lurie Children's Hospital of Chicago have partnered with families and friends around the world to produce and publish the very first edition of the **ROHHAD Reader** – a ROHHAD newsletter designed specifically for families whose everyday lives are affected by this rare disorder.

Within the pages of this **ROHHAD Reader**, you will find research updates, family stories, fun photographs, researcher spotlights, information about upcoming events and fundraising opportunities, helpful resources, shared experiences, and diverse perspectives. We hope that these pieces will help us establish open and accessible lines of communication and build a strong sense of community among the physicians, researchers, foundations, and families who are all committed to improving the lives of children and adults affected by ROHHAD.

Thank you to everyone who contributed. We look forward to growing and learning as we work to make the **ROHHAD Reader** a reliable resource for the ROHHAD community.

Very Sincerely,

The ROHHAD Reader Team

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.

A few potential symptoms of ROHHAD:

- Significant weight gain over a 6-month period in children between the ages of 1.5 and 10 years
- 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.

Currently, there is no known cause or cure for ROHHAD. However, international research teams are actively collaborating on studies that aim to increase our understanding of the disorder and how to most effectively treat and care for patients who are affected (see pgs. 13-17).

ROHHAD IN ACTION

Age: 7 **Home**: Japan
Kouya loves to play Rugby!

Age: 6 **Home**: USA
Alayna graduated from preschool!

Age: 6 **Home**: Canada
Meryem loves her dog!

Age: 9 **Home**: Malta
Jake loves to run, swim, and cycle! (Check out pg. 3.)

Age: 7 **Home**: USA
Emerson helped build a hut for his family at the beach!

Age: 10 **Home**: USA
This summer, Marky became an uncle!

Age: 13 **Home**: USA
Susanne loves to sing!

Age: 8 **Home**: Scotland
Aaron does physio in a hydrotherapy pool!

Age: 17 **Home**: USA
Trey graduated from school a year early!

PATIENT SPOTLIGHT

Meet Matthew

Age: 14 yrs Home: Texas, USA

What makes you feel strong or happy?

"...when I am able to achieve a goal that I have been working towards. The reason this makes me stronger is because even though I may have had some setbacks, I was still able to accomplish the goal with constant determination."

What do you like to do for fun?

"...drawing, playing on my Nintendo, and watching movies. I also enjoy hanging out with my friends at Dave & Busters while playing arcade games & bowling."

Who is your role model and why?

"...my mom, because she is a fighter. When I say this, I mean that whenever there has been an issue dealing with my care (doctors, school administrators, medical equipment personnel) she has always figured out some way to get what I need for my health. Sometimes it involves meetings, long phone calls, running around back & forth - whatever it takes. She has been my advocate and this is why my mom is my hero & role model."

What do you think is the most difficult part about having ROHHAD?

"...the struggle to lose weight. Even though I eat the right foods, watch my portions and go to the gym I still face the struggle. While everyone else may see that I look like I am losing weight, I don't see it in myself."

How does ROHHAD affect you at school?

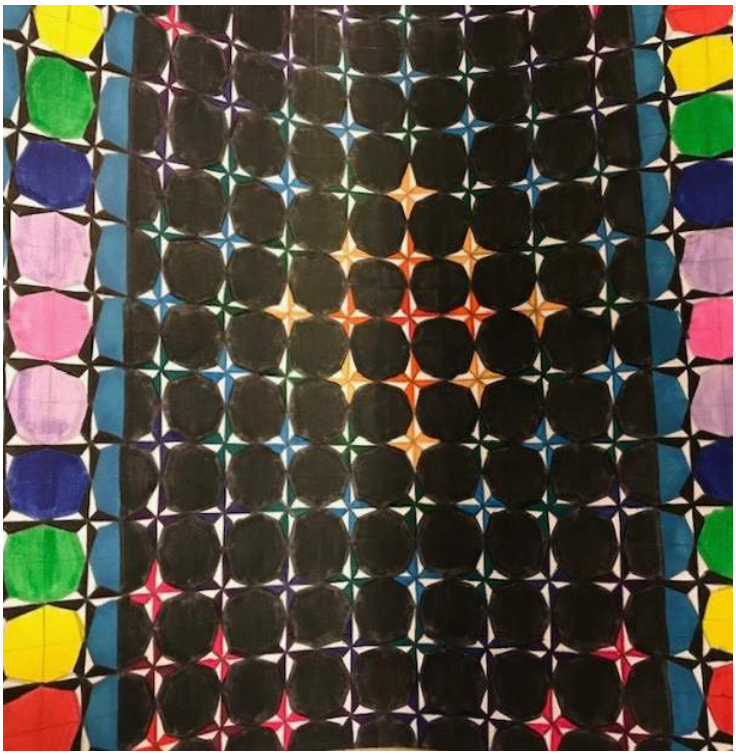
"...when temperatures reach extreme highs, my time outdoors is limited due to the problem with temperature regulation. Before ROHHAD I played soccer and T-ball without worrying about temperatures."

What advice would you give to other people with ROHHAD?

"...you need to know your body. What I mean by this, is that whenever you feel that something is not right then you should tell someone because ONLY you know your body."

A BIT OF

MATTHEW'S ART



PATIENT SPOTLIGHT

Meet Julie

Age: 27 yrs. Home: Denmark

What do you think makes you unique?

"It is my personality and my sense of humor. I have a big heart and am caring for other people."

How did ROHHAD affect you at school?

"I was in a normal school with other children. They did everything they could to make my school day as normal as possible."

What is a goal you have?

"My goal is to get up out of my wheelchair and walk all day."

What advice would you give to other people with ROHHAD?

"Love your life and fight for what you believe, no matter what others say."

What makes you feel strong or happy?

"My boyfriend, family, friends, and dog make me happy every day. They give me a smile on my face and make me laugh."

What do you think is the most difficult part about having ROHHAD?

"No one knows ROHHAD. You have to explain it again and again to new doctors and nurses."

FAMILY PERSPECTIVE

THE COOLEEN'S

Home: Maine, USA

"When Brigid was seven years old, she started inexplicably gaining weight. We initially felt that it was anxiety over our decision to move from New York to Maine but realized shortly after we moved and Brigid settled in, made new friends, and was happy, that it must be something other than stress. Brigid, by far, was our healthiest eater. We did the round of doctors and tests and everything came back negative so we finally settled in at the nutritionists. We followed her instructions to the letter.

However, by age eight, she was gaining an average of 5 pounds a month. Shortly before her ninth birthday, she had what appeared to be some sort of chronic respiratory infection. We were back and forth to the doctor's office once again. She became difficult to wake in the mornings and was constantly exhausted. Another trip to the doctor where we found that her oxygen level was 64%.

Off to the hospital! Once the battery of tests began, each result was more terrifying than the last: hypoventilation (what the heck was that?), neural crest tumor (ganglio-what?), scoliosis, diabetes insipidus (again - ?). We were absolutely devastated. No one could find a reason for of these symptoms. It drove her endocrinologist crazy because he felt that there couldn't possibly be all of these random symptoms without a connection. He found an answer! At the time, it was called Late Onset CCHS, now known as ROHHAD – thank you Dr. Olshan! He sent us to Dr. Weese-Mayer in Chicago and the fabulous team at C.A.M.P. and even though we were crushed to learn that there is no cure for ROHHAD, we were very relieved to find support, knowledge, and a plan of action to manage Brigid's condition.

It's been 16 years since our journey began, and we are profoundly grateful for all of the love and support we've found in family, friends, and the incredibly caring specialists who attend to Brigid's care." - The Cooleen Family

FAMILY PERSPECTIVE

THE POLESE-BLYTH FAMILY

Home: Belgium

What life changes has your family made to adjust to ROHHAD?

"The most significant practical changes in our family are diet and exercise. We already had a healthy diet, but are now even more aware of sugar quantities, calories and fats etc. Edwin has a pedometer watch and challenges his past records so as to be able to enjoy a richer meal. We now go on foot or by bike when the distance is under 2 km [1.2 mi] so as to get a maximum of exercise and healthy breathing into one day. He monitors his weight every morning and knows and understands what foods he can eat plenty of and what to avoid. Having our family's, the school-team and the friends' parents support is vital for this: every birthday party or excursion can mean a difference of 1 kg [2.2 lbs] which he will have to work off over a month. Fun-filled-healthy snacks make his day! As being constantly hungry is one of the ROHHAD side effects, we include Edwin in the process of shopping and cooking: he chooses recipes, helps with the groceries and loves to chop, mix and cook." - Kim Blyth (Edwin's Mom)

What have you learned from having a child with ROHHAD?

"Having a child with ROHHAD or any life-threatening disease made us realise how fragile our lives are and how blessed we are to have each other. Once we started seeing life, whatever of it was left, and not oncoming death, whenever and however it would happen, our family started living, not just again but more. We appreciate life and all the little moments, we try to spend as much time as possible as a family in activities with our relatives or close friends and even make hospital visits a fun-thing-to-do."
- Kim Blyth (Edwin's Mom)

FAMILY PERSPECTIVE

THE WARD FAMILY

Home: Colorado, USA

"27 years ago, Trevor was born a healthy, happy baby boy. Changes started happening right after his second birthday. He was in and out of hospitals for six months. Doctors could not figure out what was wrong with our first born. Trevor went into respiratory arrest at the age of 2 ½. This is when our lives changed forever. He lost oxygen to his brain for several minutes which left Trevor developmentally delayed. Trevor was never diagnosed from the beginning. Doctors could not figure out what was wrong with him. They would call it Ondine's Curse. Which just meant he did not know how to breath when he falls asleep. I hated that word! Luckily, we had one doctor who was optimistic and told us about this procedure called Diaphragmatic Pacing. Like all parents would do, we sought out to find this doctor. We were led to Dr. Weese-Mayer in Chicago. Her team started seeing Trevor at the age of 8. It was an agonizing long week of testing for Trevor. It was very emotional for me because we started to get a few answers. Answers that I did not want to hear. I was told that he needed to be on a vent for 24 hours a day and that he would never get better. Also, he was not a candidate for the Diaphragmatic Pacing. We were heartbroken! Dr. Weese-Mayer's team introduced Trevor to a new portable vent. It made our lives so much easier and more manageable. We continued to see the team in Chicago every two years and finally he got diagnosed with ROHHAD. Our last visit was in 2012. The team in Chicago was my rock for those several years. They taught me how to manage his vent so he did not need oxygen while awake. We live in Colorado so that was huge. I was able to find an adult pulmonary doc here and she would coordinate with Chicago, so we had no need to go back. As Trevor gets older, his care does not change, we just maintain his quality of life.

The hardest part of this disease is not knowing what the future brings. After a long time of hoping one day he will get better, we finally accepted that Trevor is Trevor. We love his smile and his love of life. We have learned as a family that life is a gift no matter what gets thrown at you. We live each day as it were our last. I want Trevor to enjoy and experience life to his fullest.

Trevor has two wonderful younger brothers that have been part of his care all their lives. My rock today is my family if it wasn't for my husband, sons, mom and dad and my sister, life would be extremely difficult. As Trevor gets older life doesn't get any easier because I get older. He requires 24-hour care, so our lives do revolve around Trevor. All I can say today is that I am blessed to be Trevor's mom." - Leslie Ward (Trevor's Mom)

Q&A WITH A RESEARCHER

DR. SARAH BARCLAY

Home: CANADA

Dr. Barclay recently received her PhD from the University of Calgary in Canada. Dr. Barclay's primary research focus is ROHHAD. We asked her a few questions about her work, and this is what she said:

What does a typical day look like in your lab?

"A typical day for me is usually spent on the computer, analyzing genomic data, looking for patterns in the genomes of patients, learning new analysis techniques and applying them to the data, and writing papers or grant applications. A few hours each week are spent discussing results and planning future experiments with my supervisor and our collaborators."

What is the most difficult part of your job?

"The most difficult part of my job is dealing with the frustration of failure. Each time we have thought of a hypothesis about what might be causing ROHHAD, tested it, and found that it is not the answer - knowing that patients and their families are waiting for that answer and that time is of the essence - it has been very difficult. I'm learning that part of science is understanding that each failure is still a step in the right direction as we continue to rule out possibilities and get closer to the real answer."

What is your favorite part about your job?

"My favourite part about this job is that I am constantly learning new things, which means it is never boring. I also get to work with and meet such smart and successful people, who have contributed such exciting findings to the fields I work in, so every single day I am inspired to work harder, and learn more."

What are you excited about for future ROHHAD Research?

"I am most excited about all the studies that can be done after we finally identify the cause of ROHHAD. Figuring out the cause will just be the first step, and will lead to so many exciting new discoveries about the biology of the phenotype and answers for patients and their families about exactly what is going on in their bodies. The ultimate hope is that this understanding will lead to treatment possibilities – and that is the most exciting thing of all!"

SEE PAGE 13 ! for details about one of the studies that Sarah is working on


EUIE
Home: Australia


JULIE
Home: Poland


DAMON
Home: Germany


NIENKE
Home: Europe


LEAH
Home: England

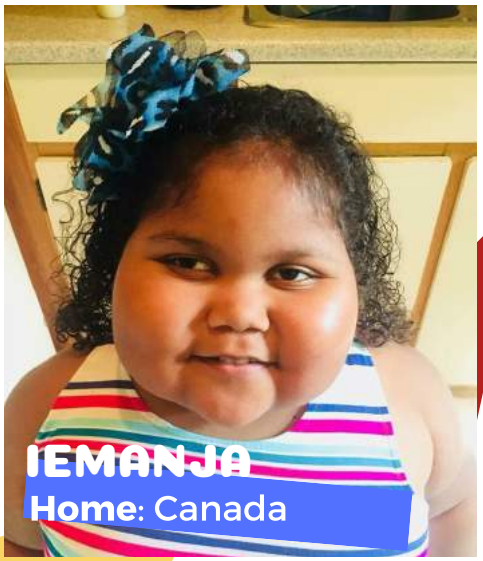

CODY
Home: USA


JAZMIN
Home: USA


SEBASTIAN
Home: Austrailia


JESSICA
Home: Ireland


CHANCE
Home: USA


IEMANJA
Home: Canada

RESEARCH UPDATE , Part 1

Biomarkers in ROHHAD

Sites: The University of Calgary, CANADA; The Ann & Robert H. Lurie Children's Hospital of Chicago, USA
Funded in part by ROHHAD Fight Inc.

A **biomarker** is any biological sign of a condition, disease, or abnormal internal process. Different biomarkers are associated with different diseases. Identifying biomarkers specific to ROHHAD would help researchers find out what causes ROHHAD and what healthcare providers can do to diagnose ROHHAD earlier and treat ROHHAD more effectively. Researchers at Ann & Robert H. Lurie Children's Hospital of Chicago have been working with Dr. Sarah Barclay (pg. 11) and a team at the University of Calgary in Canada, to look for biomarkers in ROHHAD.

So far, the research team has not found any unique biomarkers, but this study still has a long way to go. The researchers working on this project are collecting blood, saliva, and tumor samples from people with ROHHAD and their family members in order to continue the search for biomarkers of ROHHAD.

READ MORE @ https://www.luriechildrens.org/en/specialties-conditions/autonomic-medicine/research/rohhad-studies/

RESEARCH UPDATE, Part 2

Metabolic Phenotyping in Children With ROHHAD Syndrome

Site: Boston Children's Hospital, USA

Metabolism is the process our bodies use to turn food into energy. **Metabolic phenotyping** is a method scientists use to understand how metabolism is affected by our genes, environment, and lifestyle choices. Researchers at Boston Children's Hospital are using metabolic phenotyping to investigate the rapid-onset obesity that people with ROHHAD experience during childhood. The study aims to determine two primary things.

1) If changes in **energy expenditure** – the amount of energy required to carry out regular bodily function such as breathing, digestion, movement etc. – play a role in the development of ROHHAD-associated obesity.
2) If **feeding signals** – signals in the blood that tell the brain when the body is full – are dysregulated in children who have ROHHAD.

By comparing energy expenditure and feeding signals in children with and without ROHHAD, the research team hopes to provide insight into how and why rapid-onset obesity occurs in children with ROHHAD.

READ MORE @ https://clinicaltrials.gov/ct2/show/NCT02602769?cond=ROHHAD&rank=3

RESEARCH UPDATE, Part 3

The ROHHAD Registry

Site: The Ann & Robert H. Lurie Children's Hospital of Chicago, USA
Funded in part by ROHHAD Fight Inc.

The Center for Autonomic Medicine in Pediatrics has kick-started an international collaboration with ROHHAD patients and their families and physicians, in order to build an international **ROHHAD Registry.** The registry is a database of information about the health, wellbeing, and medical history of people affected by ROHHAD. The purpose of the registry is to provide clinicians and researchers with details about the health problems that people with ROHHAD experience with advancing age. The hope is to establish a better understanding of how ROHHAD affects patients over time and to help researchers identify potential causes of the disease, improve methods for diagnosis, and anticipate potential health care needs. Participation in this study involves filling out a survey on a secure web application called Research Electronic Data Capture (REDCap). If you are interested in joining this registry, you can find more information **here**.

As of July 2018, there are 83 families participating in the ROHHAD Registry!

READ MORE @ https://clinicaltrials.gov/ct2/show/NCT03135730


GENESIS


SOPHEE


BRAXTYN

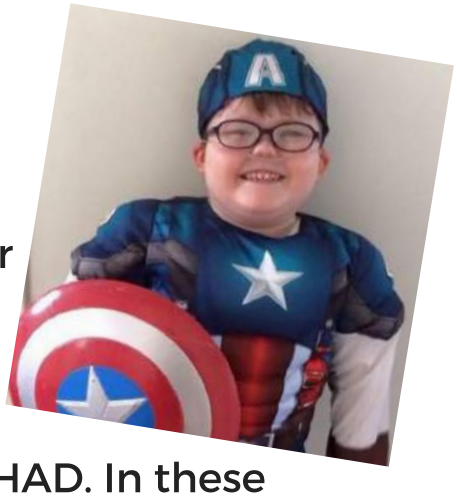
RESEARCH UPDATE, Part 4

ROHHAD EUROPEAN CONSORTIUM

Investigating Causes and Symptoms of ROHHAD

Sites: UCL Great Ormond Street Hospital Institute of Child Health (UCL GOS ICH), ENGLAND; Royal Hospital for Children, SCOTLAND; The Gaslini Institute, ITALY

Funded in part by ROHHAD Association



Researchers at UCL GOS ICH and the Gaslini Institute are collaborating to investigate potential causes and symptoms of ROHHAD. The goal of their research is to improve how ROHHAD is diagnosed and to investigate new therapies for treating ROHHAD. So far, the two institutes have separately analyzed DNA from a number of people with ROHHAD. In these analyses, both teams have identified genetic mutations that they think might play a role in the development of ROHHAD. Now, the researchers will compare the genetic mutations they have identified, in order to narrow the list of possible genetic causes for ROHHAD. They will also create models of ROHHAD in living and non-living systems in order to study the genetic mutations more carefully and to test potential new therapies for ROHHAD. Additionally, the researchers will investigate both immune function and brain structure in patients with ROHHAD, using novel tests and special imaging techniques. The study of brain structure will focus on the hypothalamus, a region of the brain that is highly suspected to be involved in ROHHAD.

Simultaneously, the European research team plans to create a sub-registry to collect information from people who are living with ROHHAD in Europe. Just like the International ROHHAD Registry (see pg. 15), this sub-registry will collect information about the current and past health of ROHHAD patients and will also include clinical, biochemical and neuroradiological data. Researchers will use information from the registry to compare individual symptoms and associated genetic mutations in order to better understand the condition and optimize care.

RESEARCH UPDATE, Part 5

ROHHAD Banking

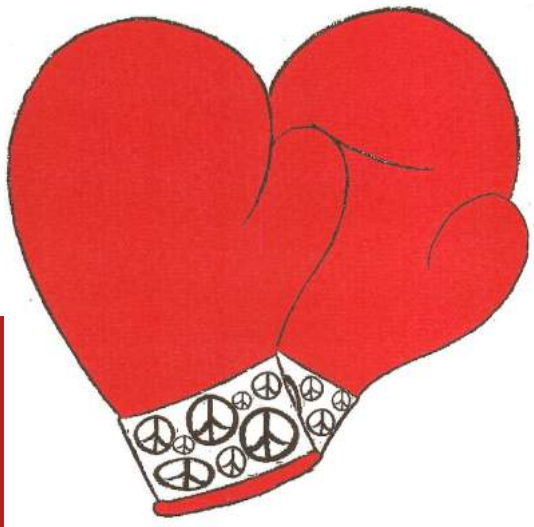
Sites: Harvard Brain Tissue Resource Center, USA
Ann & Robert H. Lurie Children's Hospital of Chicago, USA
Funded in part by ROHHAD Fight Inc.
The Harvard Brain Tissue Resource Center is funded by The National Institute of Mental Health (NIMH), the National Institute of Neurological Diseases and Stroke (NINDS), and the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD).

In collaboration with Lurie Children's Hospital and ROHHAD Fight Inc., the Harvard Brain Tissue Resource Center (HBTRC) is coordinating registration for prospective post-mortem brain donations from ROHHAD families. 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. Although we know death is a difficult thing to consider when it may be decades away, the time to start thinking about tissue 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. We encourage all ROHHAD families to carefully consider and discuss this important research.

READ MORE @ https://hbtrc.mclean.harvard.edu/
OR
https://www.luriechildrens.org/en/specialties-conditions/autonomic-medicine/research/rohhad-studies/

BUILDING COMMUNITY and RAISING AWARENESS

ROHHAD ASSOCIATION



COMMUNITY ORGANIZATIONS

A few of the community organizations supporting ROHHAD

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/

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/

UPCOMING EVENTS and ONGOING FUNDRAISERS

Research Funding

This fall, **ROHHAD Association** will open an application for funding for physicians and researchers to conduct new and continuing ROHHAD Research.

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. Schools can participate per classroom or collectively as a school. Families can participate. Check it out on the **ROHHAD Association**'s website **here**.

This year. . . be our HERO!

RACING for ROHHAD

ROHHAD Fight, Inc. is sponsoring Gary Jensen at the Ironman World Championship in **Kona, Hawaii** on October 13, 2018!

Rare Disease Day

February 28, 2019

https://www.rarediseaseday.org/

ORGANIZE YOUR OWN FUNDRAISER

You can contribute to the fight against ROHHAD! Many families have organized their own incredible events to raise awareness and funds for ROHHAD support and research. Check out the links below for ways to get started.

http://www.rohhadassociation.com/?page_id=147

https://www.luriechildrens.org/en/ways-to-help/fundraise/

https://www.flapjackfunds.com/

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.** https://smile.amazon.com/

RESOURCES

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

The ROHHAD Reader Team would like to extend a big thank you to all of the children and adults with ROHHAD, families, friends, researchers, and physicians who contributed to this very first issue of the ROHHAD Reader. **THANK YOU** for your wonderful photographs and thoughtful words. We hope you enjoyed the first final product and look forward to working with you again soon on future issues of the ROHHAD Reader.

Thank you, also, to **Danielle Carney** and **Lisa Hunter** for their invaluable consultation and to undergraduate students Juliet Torres, Lizbeth Garcia, and Roshni Patel for the contributions they made while working as summer students at Lurie Children's Hospital.

We would like to dedicate this issue of the ROHHAD Reader to the memory of **LUCA**, an incredible, 11-year-old with ROHHAD, who passed away in November of last year.

CONTACT US at ROHHADReader@gmail.com

Let us know what you thought of this first issue of the ROHHAD Reader. Send us an email with questions, concerns, comments, suggestions, or contributions for next time.