

EDITION 5

ROHHAD READER

RESEARCH UPDATES, PATIENT STORIES
&
FUN ACTIVITIES



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Welcome back!

We proudly present our 5th issue 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.

Life has changed *so much* for all of us since our last issue pre-pandemic! Routine appointments and therapies were paused indefinitely. Parents, students, and teachers have had to learn how to navigate remote learning. We can't even take getting/giving a non-virtual hug for granted!

It's hard to know when everything will go back to "*normal*." Until then it's important to take time to focus on who and what makes you happy.

Times may be tough, but the ROHHAD Reader community is tougher!

-The ROHHAD Reader Team

READER SPOTLIGHT

Troy

Age: 23

📍 Easley, SC



WHAT HAVE YOU DONE TO BEAT THE QUARANTINE BLUES?

"TROY'S OLDEST BROTHER SET UP A 'BEACH' PARTY IN HIS OWN BACKYARD BY PROVIDING PLASTIC POOLS, SPRINKLERS, CORN HOLE, BOCCI, GOOD FOOD AND BEACH MUSIC. WHERE THERE'S A WILL THERE'S A WAY!"

HAVE YOU PICKED UP ANY HOBBIES DURING QUARANTINE?

"TROY AUDITED A FOOD SCIENCE CLASS OFFERED FOR FREE, AND FOUND AN ITALIAN COOKING SHOW HE ENJOYS"

IS THERE ANYTHING ELSE YOU WOULD LIKE TO SHARE?

"TROY'S HAIR GROWS LIKE A WEED, SO OUT OF DESPERATION, HIS DAD CUT IT... WELL, TRIED TO CUT IT. HIS YOUNGER BROTHER CAME TO THE RESCUE, AND WHILE HE PLANS TO BECOME AN ARCHITECT, TATE NOW HAS BACK UP PLANS!"

WHAT ARE SOME FUN WAYS YOU HAVE KEPT IN TOUCH WITH YOUR FRIENDS AND FAMILY VIRTUALLY?

"TROY LOVES SOCIAL MEDIA, SO DOES ALL OF THAT, BUT HAS ALSO LEARNED HOW TO ZOOM GRANDPARENTS IN RETIREMENT FACILITIES."



READER SPOTLIGHT

Nina

Johannesburg, SA

Age: 21



WHAT HAVE YOU DONE TO BEAT THE QUARANTINE BLUES?

"WE [MY FAMILY] SPEND A LOT OF TIME TOGETHER AND KEEP EACH OTHER COMPANY"

HAVE YOU PICKED UP ANY HOBBIES DURING QUARANTINE?

"I LIKE TO READ AND PLAY XBOX GAMES"

WHAT WAY HAVE YOU BEEN SUPPORTED DURING QUARANTINE? ARE THERE WAYS YOU'VE BEEN ABLE TO SUPPORT OTHERS FROM HOME?

"MY FAMILY IS VERY SUPPORTIVE AND MY MOM DEALS WITH ALL MY DOCTORS AND MEDICINES"

DO YOU HAVE ANY TELEMEDICINE APPOINTMENT TIPS FOR FAMILIES PREPARING FOR THEIR FIRST ONLINE APPOINTMENT?

WRITE YOUR QUESTIONS DOWN BEFOREHAND SO YOU DON'T FORGET ANYTHING



RESEARCH STUDENT SPOTLIGHT

Every summer, the Center for Autonomic Medicine in Pediatrics (CAMP) at Ann & Robert H. Lurie Children's Hospital of Chicago mentors student researchers with the potential to make great contributions to medical research. They split their time between observing clinical activities, working on their independent research projects, and working on the ROHHAD Reader.

The ROHHAD Reader team would like to spotlight all their hard work. Thank you for being such exceptional students! You can read more about the work each student did this summer at the end of this issue of the ROHHAD Reader.



From left to right: Gabriela Martins, Nikita Menta, Remi Welbel, and Faraz Longi

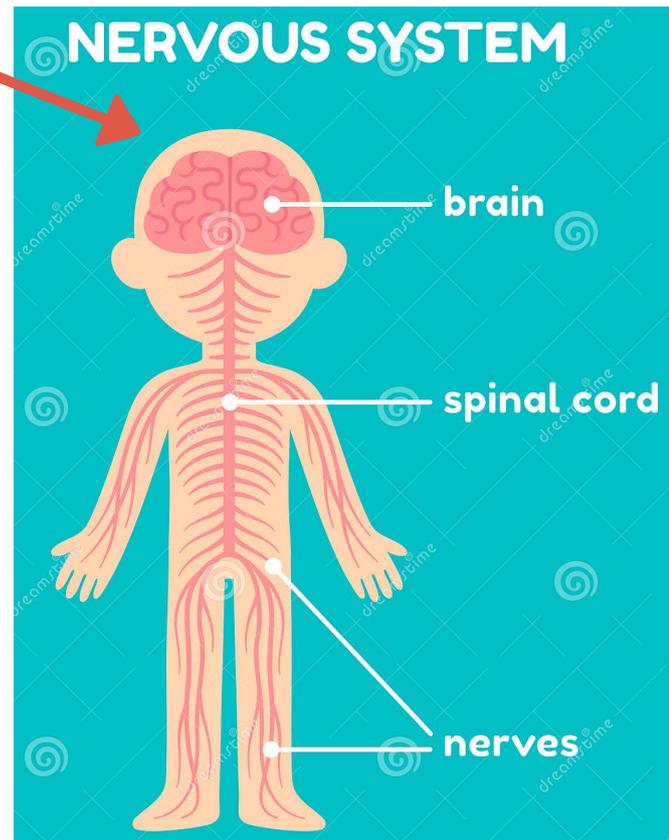
EVER WONDER WHY THE TOOTH FAIRY IS SO BUSY?

The Tooth Fairy Project!

ROHHAD affects cells from the **nervous system**. This system is made up of your brain, spinal cord, and all your nerves. Researchers have always wanted to learn what is different about cells from the nervous system in ROHHAD patients compared to people who don't have ROHHAD. But this specific type of cell has been nearly impossible to obtain for research.

Until now.....

It was recently discovered that these **cells can be obtained from teeth!** This exciting discovery could help us figure out what is causing ROHHAD. **But we need your help!**

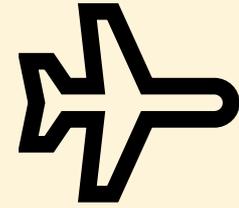
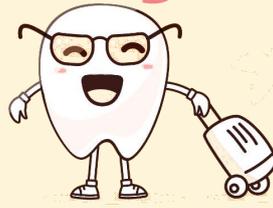
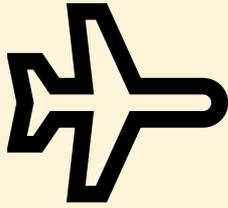


How can YOU help?

We need ROHHAD patients to donate teeth. If your child with ROHHAD will be losing baby teeth or if a ROHHAD patient will be getting any teeth removed in the future, please contact vislasmontantes@luriechildrens.org **before** the tooth is out. **This will allow us time to mail out a tooth collection kit to you ahead of time.**



KOUYA'S TOOTH: AMAZING JOURNEY!



Kouya lives in Japan and has ROHHAD. He is one of our favorite ROHHAD Reader contributors. This is the story of the amazing journey of Kouya's tooth all the way around the world and what it is doing to help ROHHAD research.

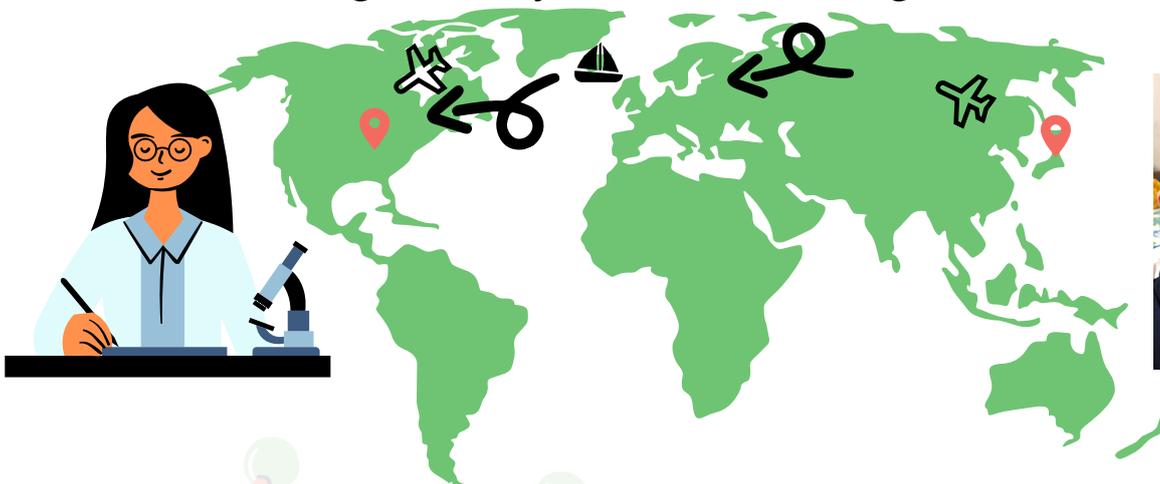


Kouya had consented to participate in the Tooth Fairy project and had received the special tube to store and ship his tooth in when it came out. But, when it finally came out, restrictions due to COVID-19 made it nearly impossible to ship the tooth to our Tooth Fairy collaborator in the USA.

Kouya and his family kept the tooth in the tube safely refrigerated for almost **FOUR** weeks before the tooth could begin its **6,600 or 10,584 km journey** to our laboratory in the USA.

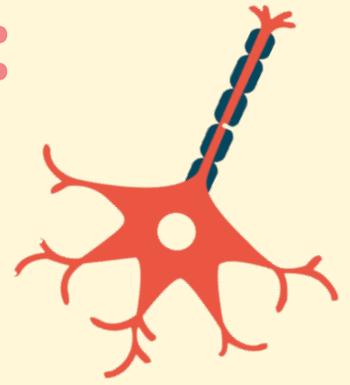
Kouya's tooth safely arrived after its LONG journey to the Tooth Fairy project lab at the University of Tennessee...but was it too late or could the scientists still use the tooth to help ROHHAD research? Keep reading to find out!

Thank you to the Hashimoto family for their dedication to making sure Kouya's tooth donation got to the lab!





KOUYA'S TOOTH: FROM TOOTH TO NEURON!



When Kouya's tooth finally arrived, scientists were ready!

When we lose a tooth, there is a special type of cell that comes out with the tooth. These cells are known as **dental pulp stem cells (DPSC)**.

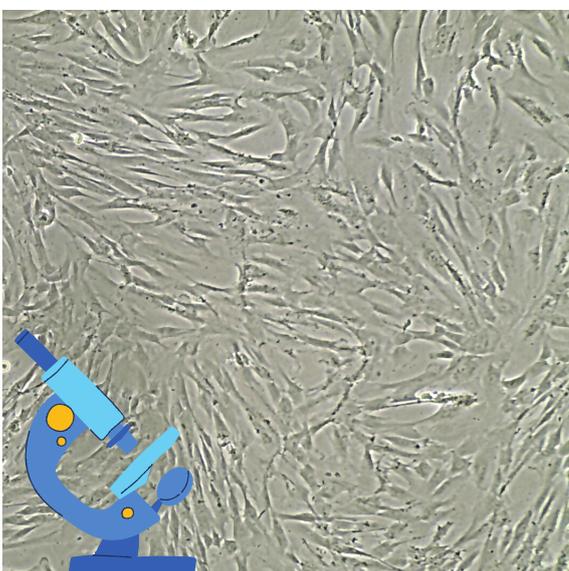
Scientists can use these special cells to develop many different type of cells. For ROHHAD, scientist hope to create **neurons**. **Neurons** are cells from your nervous system, the cells scientist think are affected in ROHHAD!

DPSCs from Kouya's tooth went through a special process and.....**success!** Scientists were able to create **neurons** from these DPSC. **Amazing!**
See the pictures below!

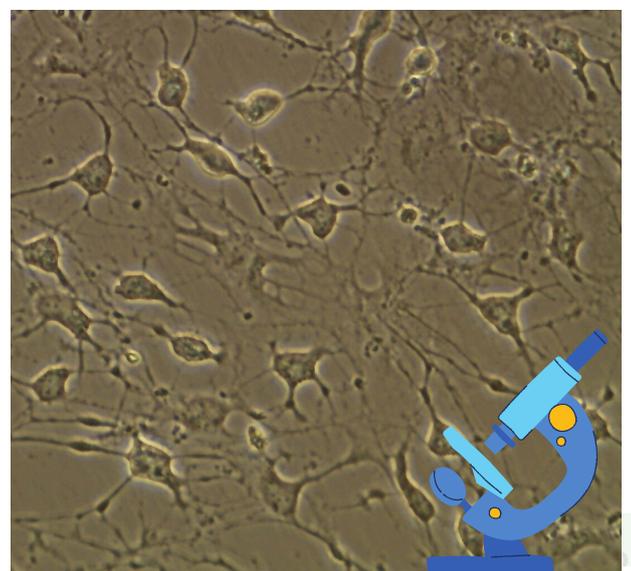
Thanks to Kouya and our other Tooth Fairy Project participants, scientists will be able to look closely at how neurons are affected in ROHHAD and better understand what we need to do to treat ROHHAD.

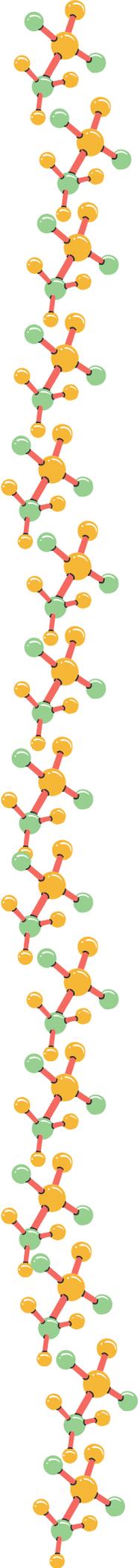
Email Val at vislasmontantes@luriechildrens.org if you want to participate too!

Dental Pulp Stem Cells



Neurons

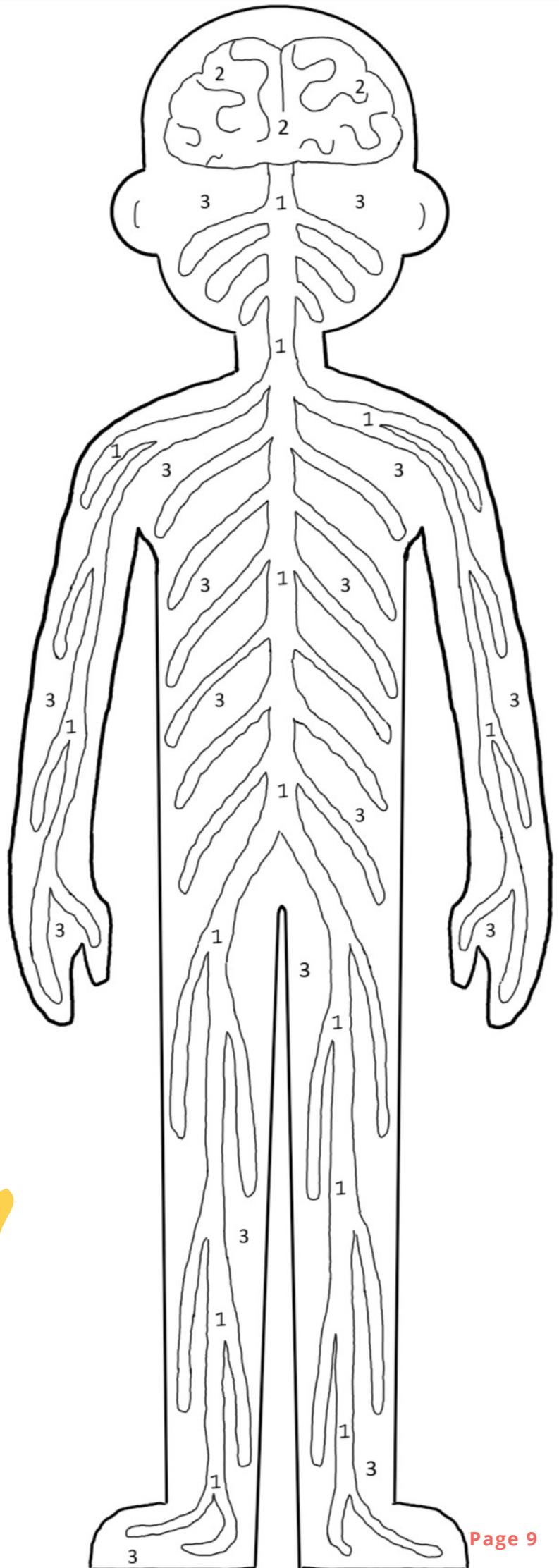
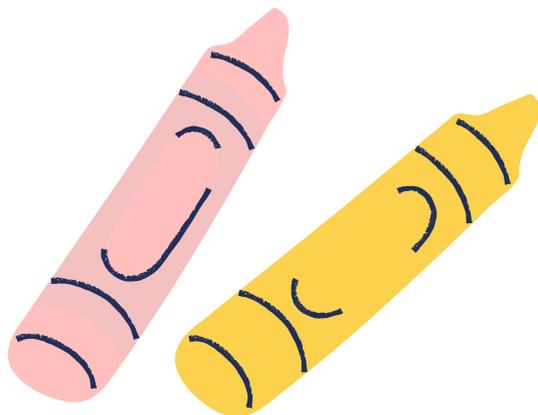




COLOR BY NUMBER - NERVOUS SYSTEM

Yellow = 1
Pink = 2
Skin Color = 3

The nervous system controls everything you do, like breathing, walking, and thinking. The nerves live all over the body. This is how your brain and the rest of your body communicates.



ROHHAD REGISTRY UPDATES



63

Patients enrolled in
the International
ROHHAD Registry!



Since July 2019, we have had 13 new participants join the Registry! Great work ROHHAD community!

The Registry is used by a collaborative team of physicians, scientists and researchers around the world who want to learn more about ROHHAD so they can find better ways to treat it. It uses **REDCap**, which is a very secure system designed to keep health information safe.



If you are not part of the Registry yet, **[please click here](#)** or go to the link below to the process started.



<https://redcap.link/zzue68pv>

If you are already in the Registry but have not updated your information recently, please reach out to the Registry team at **CAMPResearch@LurieChildrens.org**

GENOMICS AND GENOME SEQUENCING



DNA is a molecule that carries information about how we look and function. Many DNA molecules are used to make up genes. Genes are like "**blueprints**" that tell a cell how to make **proteins**. These proteins determine how the cell looks and works. Genes determine many of the traits that we inherit, like eye color and hair color. They also play a big role in our health.

These DNA "**blueprints**" are written in a special letter code called the genetic code. The four letters of the genetic code are A, T, C, and G. They are grouped into triplets (groups of three) that code for the building blocks of **proteins**, called **amino acids**.

Word Bank:

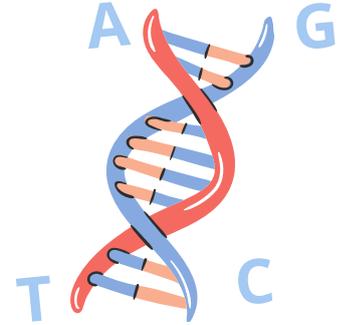
Protein: proteins are naturally found in plants and animals. There are in almost every part of your body, like your hair, muscles, bones, and skin!

Amino acids: amino acids are the building blocks of proteins. They get strung together like beads on a string.

GENOMICS AND GENOME SEQUENCING



WHAT IS GENOMICS?



When there are mistakes in the **"blueprint"**, the proteins that our bodies need might not be built properly or at all. By **sequencing** a person's genetic code, scientists can investigate **mutations** in their genes that may put them at risk for disease.

By gaining an understanding of a disease at the **"blueprint"** level, scientists can develop new treatments. This is why genomic sequencing is a promising tool for learning more about the causes of ROHHAD and finding ways to treat it!

Word Bank:

Sequencing: figuring out the sequence of the four repeating letters that make up the DNA molecule.

Mutation: a mistake in the genetic code or "blueprint".



CAN YOU DECODE THIS DNA SEQUENCE?



DECODER BANK:

CTT = L

AAA = Y

GGG = A

GTC = E

ACC = U

GCC = R

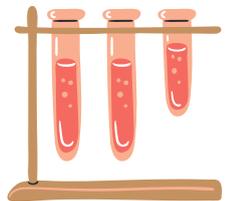
ATG = O

GAG = B

CAC = T

CCC = I

TAG = F



*Use the decoder bank to help the scientist
decode the message on the next page!*

CAN YOU DECODE THIS DNA SEQUENCE?

Hint: Match each triplet to the letter given in the decoder (for example, **AAA** equals the letter **Y** according to the decoder)

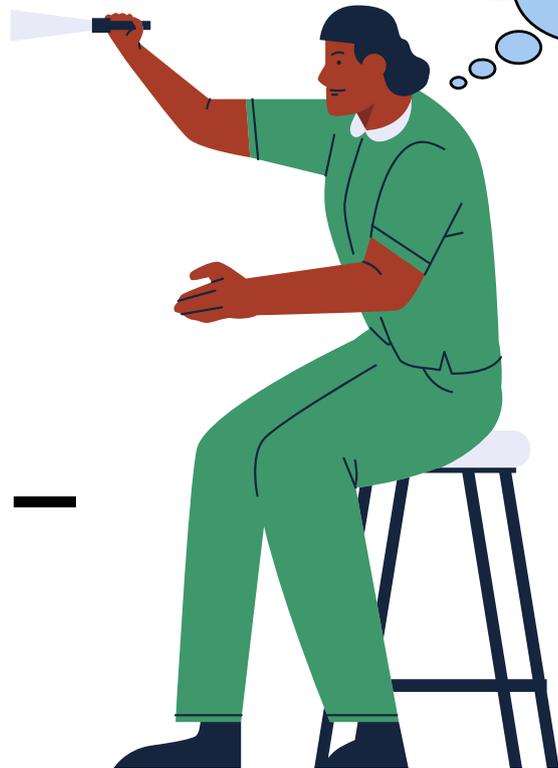
AAA **ATG** **ACC**

GGG **GCC** **GTC**

GAG **GTC** **GGG** **ACC** **CAC** **CCC** **TAG** **ACC** **CTT**

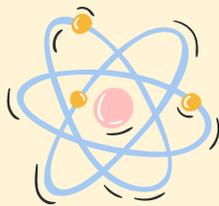
Y _ _

_ _ _



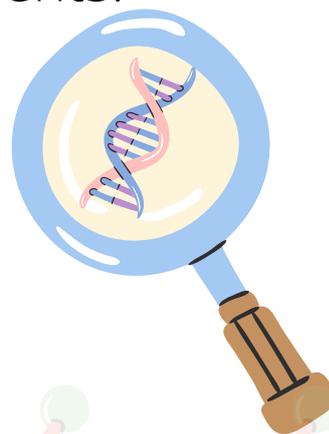
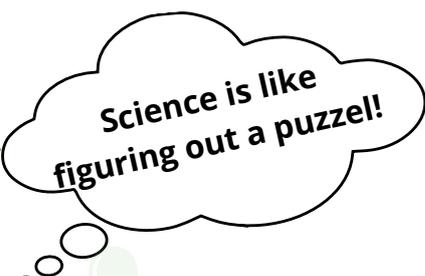
Put a decoded letter on each line!

WHAT GENOMICS RESEARCH IS HAPPENING NOW?

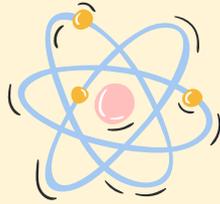


Scientists from all over the world are working together to identify gene(s) or biomarkers which may contribute to or cause ROHHAD. The purpose of these studies is to gain a better understanding what causes ROHHAD.

By sequencing the DNA of people with ROHHAD, the researchers are able to learn more about what is happening at the **"blueprint"** level, and get closer to understanding the causes of ROHHAD. This project will go a long way toward improving diagnosis and care of ROHHAD patients.



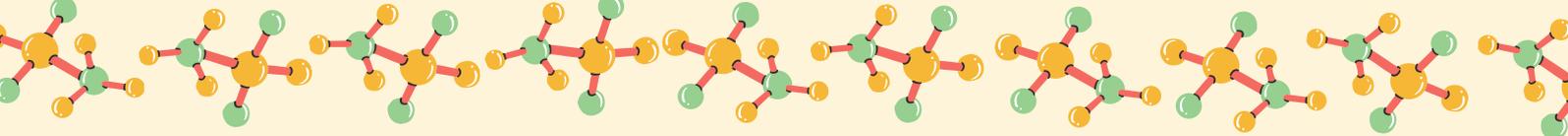
WHAT GENOMICS RESEARCH IS HAPPENING NOW?



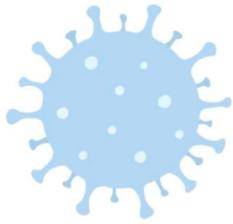
Scientists who are now studying ROHHAD previously discovered that mutations in a gene called *PHOX2B* cause a disorder named CCHS. This discovery allowed for earlier diagnosis, improved treatment, and clinical trials to improve outcomes in CCHS.

ROHHAD and CCHS have many overlapping features, so these scientists hope to make a similar discovery in ROHHAD. The genes responsible for ROHHAD remain unknown right now, but genomic sequencing and other research tools are helping scientists investigate potential genes and biomarkers. You can read more about how you can participate in some of this genomics research by clicking [here](#) and [here](#)!





COVID-19 GUIDELINES



What is COVID-19?

Coronavirus is a type of **virus**, and COVID-19 is a new type of coronavirus. Viruses are very very tiny so you can't see them, but they can make people sick

How is COVID-19 spread?



Coronavirus is spread by coughing, sneezing, and even talking (especially within 6ft or 1.8m of each other) and close personal contact

What are the symptoms?



Symptoms of confirmed COVID-19 patients include cough, fever, shortness of breath, and general flu like symptoms. Some patients get really sick and need to go to the hospital

DO

Always wear your mask, especially when in crowded areas



Maintain a distance of 6ft or 1.8m from others when in public



Wash hands and use hand sanitizer **frequently**, especially before eating or touching your face

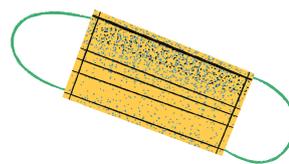


DO NOT

Touch the fabric of your mask (especially the side you breathe from) or your face

Travel if you have any symptoms or if you've been in contact with a symptomatic or asymptomatic COVID-positive person

Panic. If you're taking proper precautions then your risk of getting the virus is reduced





MORE ROHHAD RESEARCH OPPORTUNITIES

Researchers in Europe are teaming up to investigate ROHHAD. They have identified genetic changes in ROHHAD patients that may play a role in ROHHAD, and are collaborating to study these.

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.

Please contact Professor Mehul Dattani and his team at the UCL GOS Institute of Child Health (m.dattani@ucl.ac.uk)!





INVESTIGATING NEUROINFLAMMATION: THE BOSTON CHILDREN'S HOSPITAL STUDY

Researchers at Boston Children's Hospital are in the process of exploring the possible neuroinflammatory causes of ROHHAD Syndrome.

To date, they have enrolled over fifteen patients with ROHHAD for a study examining the role of inflammation and hormones in this condition, and enrollment is ongoing.

Participation can occur remotely, there is no travel required to take part.

Please contact the research coordinator for this study at **rohhad@childrens.harvard.edu** for more information!

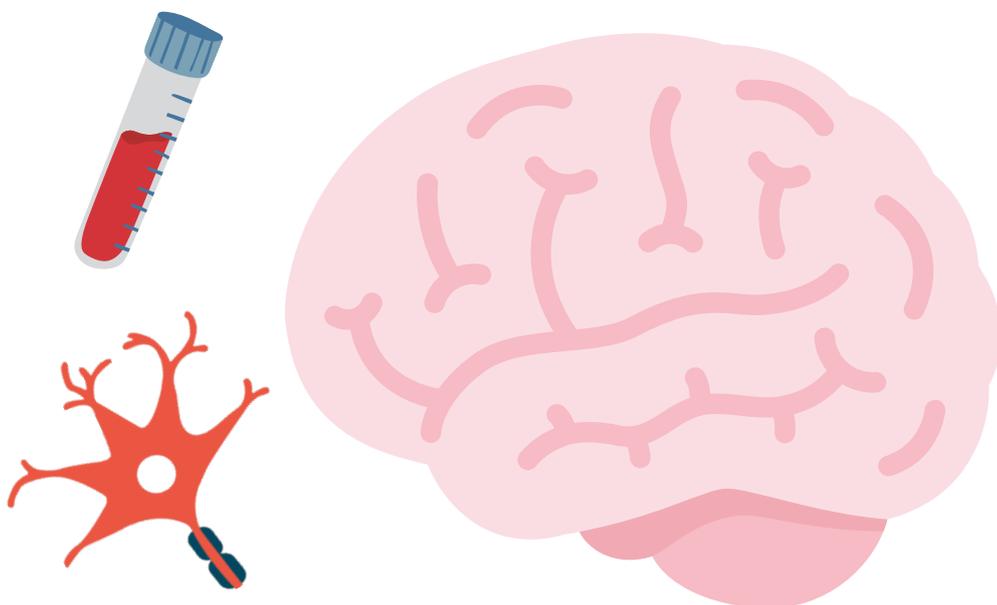


INTEGRATED WHOLE GENOME AND TRANSCRIPTOME SEQUENCING

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, thirst, hunger, and hormones.

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. The nervous system includes your brain and spinal cord.

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



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 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



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/>



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 @ www.rohhadssyndromesecretariatinjapan.netcommons.ac

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

Buy charity merchandise in support of the ROHHAD Association!

To place an order email:

ROHHADAssociation@gmail.com

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](#).

RESEARCH STUDENT SPOTLIGHT

Remi  Chicago, IL

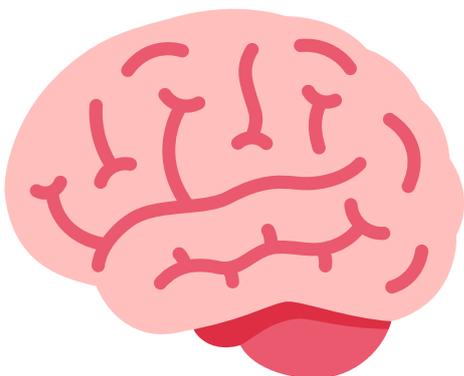


Hello! My name is Remi Welbel. I'm from Chicago and I'm a junior at Middlebury College majoring in neuroscience and minoring in dance.

I spent this summer conducting research on CCHS and ROHHAD at the Center for Autonomic Medicine in Pediatrics under the mentorship of Dr. Weese-Mayer.

My independent research project focuses on how these children learn and grow. Children with CCHS, like children with ROHHAD, can experience moments when their brain is not getting enough oxygen. To prevent this from happening, children with CCHS and ROHHAD receive treatments to help them breathe (such as ventilation through mask or tracheostomy). My project used a special game-based iPad app to collect data on how CCHS and ROHHAD patients think and learn. By looking at the relationship between the breathing treatment a child receives and their scores on the app, I will be able to see if certain treatments are associated with better scores. I hope that my research promotes earlier and more effective breathing support that enhances the mental, physical, and social well-being of these children.

In my spare time, I enjoy cooking, hiking, hammocking on Lake Michigan with my friends, and volunteering as a reading mentor for children.



ROHHAD Reader, Issue 5



RESEARCH STUDENT SPOTLIGHT

Nikita 📍 Chicago, IL

Hi! My name is Nikita and I grew up in the suburbs of Chicago. This year I am a senior at Northwestern University, where I am double majoring in communication sciences and disorders and neuroscience.

This summer I worked on a project with Faraz, another CAMP summer research student. Our project looked into peripheral skin temperature - the skin temperature of different spots on your hands and feet. We

wanted to figure out how healthy children's skin temperature changes as they get older. Does it get hotter or colder?

We also wanted to see if children with ROHHAD and CCHS have similar or different skin temperatures compared to those without these diseases. How does the skin temperature of children with ROHHAD and CCHS change as they get older?



The red dots show some of the places we collected temperatures from!



In my free time I love playing golf with my friends and family, practicing dance, and watching movies!

RESEARCH STUDENT SPOTLIGHT

Faraz  Chicago, IL

Hello! My name is Faraz Longi and I'm from the suburbs of Chicago. I'm currently a senior at the University of Michigan majoring in Biomolecular Science and I have a passion for clinical research!



The thermometer used to take peripheral skin temperature values

The autonomic nervous system controls many functions for our body including our breathing and skin temperature. This summer, I worked with Nikita to assess how peripheral skin temperature changes in patients as they get older. By looking at the peripheral skin temperature in ROHHAD, we hope to provide insight to clinicians that may allow them to make earlier diagnoses!



In my free time, I enjoy playing tennis, cutting my friends' hair, and reading!

RESEARCH STUDENT SPOTLIGHT

Gabriela  New York, NY

Hi, my name is Gabriela. I am a sophomore studying electrical engineering and computer science at New York Institute of Technology. After I graduate, I plan to do a masters. I want to work in research and design of medical devices and sensors.

ROHHAD patients cannot breathe on their own while asleep and even sometimes while they are awake. To allow kids to move around more easily, a great alternative to a mechanical ventilator is a respiratory pacemaker. It is similar to a heart pacemaker, except it stimulates breathing rather than heart rhythm. Like all technology, over time pacemaker components can break and stop working. It is important to be able to find a way to predict when this will happen to properly take care of the issue beforehand.

When ROHHAD patients visit CAMP, data are recorded about how a patient's respiratory pacemaker is functioning. My research investigates these data to see if there is information that might help us predict when a pacemaker will need to be serviced or replaced. I hope this research can help improve the safety of these patients.

