KAT6A News

Volume 4 | April 2019 | KAT6A Foundation, Inc. | www.kat6a.org

The KAT6A PATIENT REGISTRY launched on January 8, 2019!

To date we have 38 families registered. Please take the time to complete this crucial research tool as soon as possible. Researchers are depending on this data.
http://www.kat6a.org/kat6a-registry/

Read the full press release on page 7.

NORD 2019 MEMBER

Inside This Issue

PG. 2-4
Events Recap

PG. 5-6
Fundraising and Future Events

PG.7-8
Research

PG.9
Family Spotlight

PG.10
Community Highlights

PG.11
Who We Are

KAT6A Clinic 2019

The second KAT6A clinic took place at the Kennedy Krieger Institute – Johns Hopkins Medical Institution, Baltimore, Maryland, USA on February 9, 2019. Forty KAT6A families were in attendance.

The day started with Peter Najm, who is a 10 year old boy affected by KAT6A Syndrome. Peter is nonverbal, but he recently started singing. Peter sang and played on the piano the American National Anthem.

Dr. Jacqueline Harris, Assistant Professor of Neurology and Pediatrics at Johns Hopkins Medical Institution, spoke about cognitive phenotyping in KAT6A and other Mendelian disorders of epigenetic machinery. She presented a pilot study that consists of collecting clinical data and validated parent questionnaires in the hope that the data will lead to the design of a larger study to look at specific cognitive phenotypes. Most of the families attending the clinic signed the consent to participate in the pilot study. The study is open for all families wishing to participate. Please contact Natacha Esber for information on how to enroll in this study.

Dr. Anne Voss, Associate Professor at Walter and Eliza Hall Institute, traveled all the way from Australia to share her work on KAT6A with us. Her presentation was a summary of the roles of KAT6A and related proteins in mouse models of human embryonic development and disease, she also showed the methods used to test potential treatments in the mouse model.

Dr. Xiang-Jiao Yang, Professor at the Department of Medicine and Division of Experimental Medicine at McGill University came from Canada and he shared his extensive experience studying human histone acetyltransferases and focused mainly on KAT6A (a.k.a MOZ).The highlight of his presentation was his advice to check for special abilities or talents and focus on developing those in patients affected with the KAT6A gene mutation.

Dr. Richard Kelley, former Associate Professor of Pediatrics at Johns Hopkins Medical Institution, former Director of Division of Metabolism at Kennedy Krieger Institute and current researcher at the Division of Genetics at Boston Children's Hospital, presented evidence of mitochondrial disease in KAT6A syndrome after studying multiple amino acid levels and other metabolic testings in affected individuals. He showed the benefit of a mitochondrial treatment in KAT6A syndrome and the role of Tricitrates in the gastro-intestinal tract system. His main goal is to continue to gather metabolic data to guide treatment.

In the afternoon, KAT6A Foundation leaders spoke about the accomplishments of 2018 and future ambitions. The survey response to the event was overwhelmingly positive. Families enjoyed coming together to meet in person and to learn more about how research is moving along into our very rare disorder. We will consider all feedback when planning an even better clinic for 2020. Special thanks to all the volunteers who made this event a huge success. We couldn't have done it without you!

Video of the speakers and their slides are available in the KAT6A Support Group.

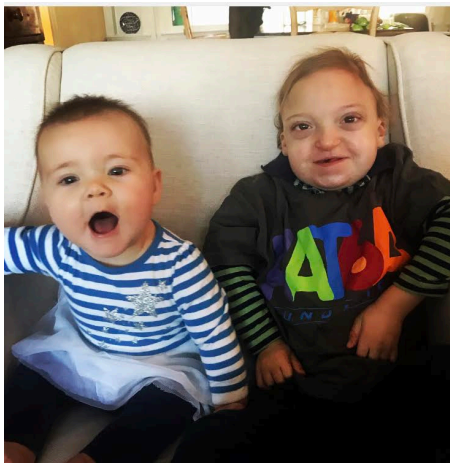
Rare Disease Day

In the US, a rare disease is qualified as one that affects as little as 200, 000 people. In Europe, it is defined as a disease that affects up to 1 in 2,000 people. Currently there are approximately 200 known KAT6A cases. We are RARE.

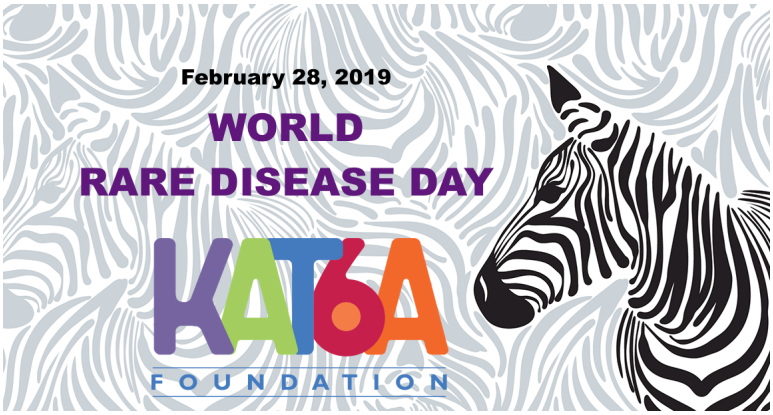
In honor of Rare Disease Day, several families raised KAT6A awareness in school. Will and Izzy had KAT6A celebrations in their classrooms. Their classmates wore stripes to show their support for all individuals with rare diseases. Will's class made a bulletin board for him, created a zebra craft, ate zebra cakes and Will even got to lead a parade around his school.

#ShowYourStripes

Look at our KAT6A kiddos and their siblings raising awareness. Our support group members plastered Facebook with the KAT6A photo frame for the month of February to honor Rare Disease Day.



In the Press



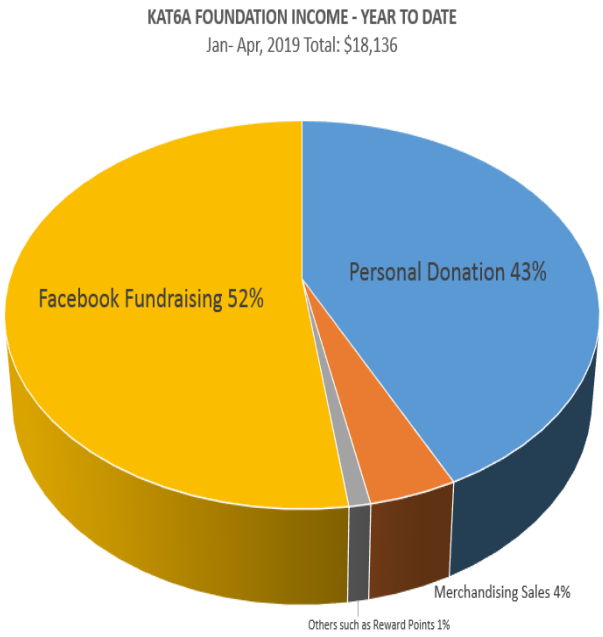
In Canada, Parker and his parents Amber Moore and Ryan Brown were featured in their local newspaper for Rare Disease Day. Amber says: "We've never really looked at Parker as a child with a rare disease. He is a unique child with some unique needs, but Parker is Parker. And that is pretty great!" Read the full article at **https://barrie360.com/local-parents-celebrate-world-rare-disease-day/?fbclid=IwAR209EN9YIbGdLq9KoRM0lZj-z993x7V_HnmXn1fpmMeQhfd-RATRaqKsoc**

Meg Salisbury was very busy on Rare Disease Day in Australia. Meg paid a visit to the breakfast radio show at Hit 104.9 to speak with Riley-Rose about KAT6A and the challenges faced by those with a rare disease. Meg and Riley-Rose showed support to the rare disease catch cry to 'wear your stripes' and added their own unique stripes to their faces.

Meg then visited Ginger's new school to speak to the entire primary school about KAT6A. All the teachers and principal wore stripes and Ginger's classmates decorated their faces with stripes. The students were then given a zebra to decorate in their own way to show that we are all different. This day had a huge impact on the students with a lot of positive feedback from parents. The students now have a greater understanding of Ginger's communication style which has extended to a much more inclusive and supportive environment both in the classroom and in the playground. In short, Ginger has become the 'rockstar' of her school and is loved by all.

Check out the video Ian Mendel put together for RDD on our Youtube channel. **https://www.youtube.com/watch?v=zL83t4GNwjg&t=45s**

FUNDRAISING



Earnings News

The KAT6A Foundation was able to raise over $400 at the clinic in February through a raffle. There were 8 lucky winners of some great KAT6A items from our official KAT6A shop.

The KAT6A Bonfire t-shirt fundraiser raised over $450. Please post photos with your KAT6A STRONG shirts on Facebook or Instagram!

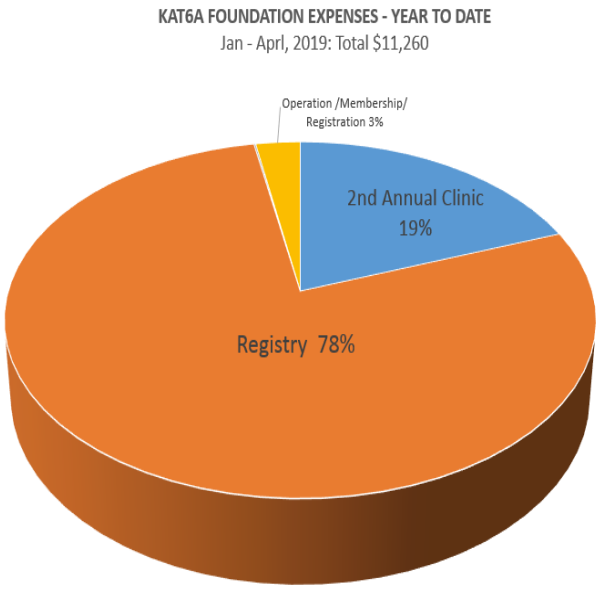
A special thanks to the following individuals for showing support in 2019 via **Facebook fundraisers**: Marc Monso Cairol, Audrey Domsten, Miri Duston, Sonia Gates, Matt Goes, Olya Good, Rui Grosa, Noe Sanchez Espinola, Tony McKinlay, Patricia Richmond, and Jeanne Syverson.

Ways to Support KAT6A Fundraising:

- Hold a Birthday Fundraiser on Facebook
- Shop using smile.amazon.com
- Purchase KAT6A awareness clothing and accessories from our shop at **http://www.kat6a.org/shop/**
- Share our website donation page
- Ask your company if they would consider donating to the KAT6A Foundation
- Hold a bake sale or lemonade stand
- Spread awareness on social media
- Join the **KAT6A Foundation Fundraising and Awareness Group** on Facebook.
- Contact Jessica Vogland to join the KAT6A Foundation Fundraising committee at **jljohnson929@yahoo.com**

Coming soon!

We are currently designing an official KAT6A awareness ribbon. Stay tuned! We will post the image as soon as it's finalized.



EVENTS

SAVE THESE DATES:

KAT6A Walk to Find a Way

September 14, 2019

Walks will take place in North Carolina and Connecticut. We are hoping to add a couple other locations, so please contact Jessica Vogland at **jljohnson929@yahoo.com** if you are interested in holding one in your city. If you cannot make one of the official walks, please consider hosting a virtual walk with your friends and family around your own neighborhood. This is an international event and we hope to increase participation this year. Last year we raised over $25,000 and we hope to double that toal in 2019. More details about the second annual walk and how to sign up will be coming soon. This information will be shared in our Facebook support group as soon as it becomes available.

KAT6A Clinic 2020

March 28, 2020

We are proud of the partnership we have formed with the medical doctors at the Kennedy Kreiger Institute and are gracious for their continued support and look forward to joining together again in Baltimore, MD, USA for the third KAT6A Clinic. Dr. Harris and Dr. Fahrner will hold appointments on March 27 for KAT6A families.

200 KNOWN CASES

VOLUNTEERS NEEDED!

We are looking for individuals to assist in collecting data for the **KAT6A registry**. Training will be provided. Please contact Emile Najm at **kat6a@yahoo.com** to get involved.

If you are interested in writing a blog post for the webpage, please contact Aimee Reitzen at **kat6aimee@gmail.com.**

Please like/follow/subscribe to us.

KAT6A Foundation

officialkat6a

NEW!!

KAT6A Foundation

KAT6A Foundation

SUPPLIES NEEDED!

We are compiling a list of durable medical devices and gently used speech aids that your children have outgrown so that these items can be passed on to other KAT6A individuals. Please contact Lindsey Geiger via Facebook messenger if you can help support this cause.

RESEARCH

KAT6A Registry

The KAT6A Foundation and NORD Launch Natural History Study of KAT6A Syndrome

Research study is open to participants worldwide to advance understanding and treatments for rare disease causing KAT6A Syndrome

West Nyack, NY, [January 08, 2019]—The KAT6A Foundation and the National Organization for Rare Disorders today launched the largest-ever study to research KAT6A Gene Mutation that causes KAT6A Syndrome. KAT6A Syndrome currently has no cure.

The new study, KAT6A Patient Registry creates a platform for patients around the world to share information about KAT6A Syndrome. Its purpose is to build an international resource to be used by scientists in future research.

"The KAT6A Patient Registry will provide a complete picture of each patient's experience with KAT6A syndrome," said KAT6A Foundation Chief Executive Officer Emile Najm. "We are launching this initiative to help fill the missing link researchers and medical experts need to advance research and get to a cure."

To help drive awareness and participation, The KAT6A Foundation will highlight information about the registry on its social media (Facebook, website, Instagram and YouTube).

"Our goal is to enroll as many patients, or their parents or legal guardians, as possible," said Najm. "The success of the registry is dependent upon community participation."

The KAT6A Patient Registry is a natural history study that consists of electronic surveys to collect information about the patient experience and disease progression. Patients, or their caregivers or guardians, can enter information from anywhere in the world. The data is made anonymous and stored securely in an online portal called a registry. The KAT6A Foundation may share the data with individuals or institutions conducting research or clinical trials, as approved by the study's governing board that includes scientists, doctors and patient advocates.

The KAT6A Foundation is launching the study in collaboration with the National Organization for Rare Disorders (NORD), an independent charity that built its natural history study platform as part of its mission to help identify and treat all 7,000 rare diseases.

The KAT6A Foundation is a member of NORD and the organizations work together to eliminate the challenges that rare disease patients face.

"Patient-powered registries are changing the landscape of rare disease research," said Vanessa Boulanger, NORD's Director of Research Programs. "By building strong partnerships within the community and with leading scientific experts, NORD's Registry Program is well-positioned to address knowledge gaps and accelerate the development of discoveries that save lives. We are so pleased to welcome The KAT6A Foundation, as a partner in our IAMRARE™ Registry Community!"

The KAT6A Syndrome is a rare genetic disorder that occurs in about 150 known cases worldwide people. Common traits are: developmental delay, intellectual disability, feeding difficulties, constipation, acid reflux, significant speech and language deficits, heart defects, seizure disorders, frequent infections, sleep disturbances, abnormal muscle tone, vision problems, behavioral challenges, small head size and distinct facial features.

For more information, visit
http://www.kat6a.org/kat6a-registry/

About National Organization for Rare Disorders (NORD)

The National Organization for Rare Disorders (NORD)® is the leading independent advocacy organization representing all patients and families affected by rare diseases. NORD is committed to the identification, treatment and cure of the 7,000 rare diseases that affect 30 million Americans, or 1 in every 10 people. NORD began as a small group of patient advocates that formed a coalition to unify and mobilize support to pass the Orphan Drug Act of 1983. For more than 30 years, NORD has led the way in voicing the needs of the rare disease community, driving supportive policies and education, advancing medical research, and providing patient and family services for those who need them most. NORD represents more than 250 disease-specific member organizations and their communities and collaborates with many other organizations in specific causes of importance to the rare disease patient community.

Media Contact: For The KAT6A Foundation: Emile Najm, 845-282-4979, **kat6aregistry@gmail.com**, For NORD: Laura Mullen, **lmullen@rarediseases.org**

NEW!!

Research into Speech & Language Skills in Individuals with KAT6A/B Variant: An International Study

Meg Salisbury in Australia spoke to Channel 9 news about the launch of the Patient Registry. The story was seen by Prof. Angela Morgan, the leader of The Centre of Research Excellence in Speech and Language at the Murdoch Children's Research Institute. Angela contacted Meg to discuss the possibility of conducting research in individuals with KAT6A variants and to access the Patient Registry. For anyone interested in participating in this research, please email **angela.morgan@mcri.edu.au**.

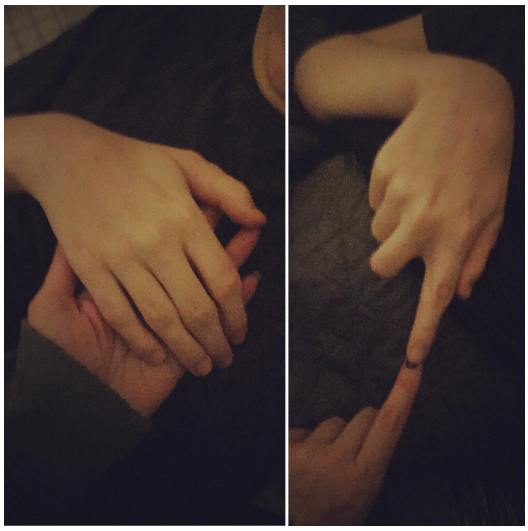
The Centre of Research Excellence in Speech and Language is an international collaboration of experts in the fields of speech pathology, paediatric neurology, neuroscience, genetics and bioinformatics whose core vision is to transform speech pathology practice by identifying, understanding and targeting the underlying causes of developmental speech and language disorders. The Centre of Research Excellence in Speech and Language is funded by the National Health and Medical Research Council for the next four years.

Connecting Families Around the World

Today we know that there are at least 200 individuals diagnosed with KAT6A. KAT6A families are living in over 28 different countries including: Argentina, Australia, Austria, Belgium, Canada, Chile, China, Denmark, Dominican Republic, England, Finland, France, Germany, Ireland, Israel, Japan, Lebanon, Netherlands, New Zealand, Norway, Portugal, Qatar, Scotland, Spain, Sweden, Turkey, United States and Venezuela. Chani Honeyman has been helping connect KAT6A families around the world with an interactive map that she created using Google Maps. Contact Chani in our Facebook KAT6A support group if you would like your family to be added to the map.

What Mommomm Sees

by Sonia Gates



These beautiful hands belong to my favorite person in the world.

What a geneticist sees

Markers for a genetic anomaly, including long, thin fingers, extended webbing between the index and middle fingers, and very deep thumb creases. We now know that these are common markers for KAT6A Syndrome.

What I see

Hands that are sweet and gentle, that frequently grab my own hands and put them up on his soft head for comfort. I see hands that navigate an iPad screen at lightning speed, to find the next elevator video, or blow stuff up with TNT on Minecraft. Long, thin fingers that sign things like "drink," "eat", and "you." In the second frame, he is pointing his index finger toward mine, while saying, "Oooooo!", which is Braedenese for "I love you!" With this new diagnosis and all of the new information I'm learning, it's easy to get caught up in the clinical lingo and the sterile dialogue. I want to always remember that first, he is my boy. The love of my life. No matter what new information we learn, he is a loving, loved, sweet, wonderful, perfect, sometimes anxious, sometimes naughty, human being. Not a diagnosis. Not a statistic. I'm lucky I get to hold those beautiful hands each night while he still lets me tuck him in.

Sonia is the mother of Braeden, age 18, recently diagnosed with KAT6A Syndrome. Braeden's symptoms include: Arnold-Chiari malformation, apraxia, orchiopexy repair, sensory issues, failure to thrive, low muscle tone, and acid reflux.

Read more from Sonia at ***www.kat6a.org/2019/4/11/18-years-and-31-days/***

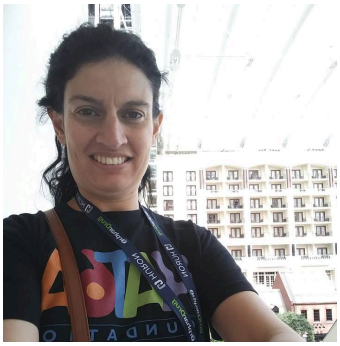
KAT6A Community Highlights

Look at these smiles! From L to R, are Mady, Hudson, Cannon, and Gianna meeting up in Austin, TX.

In addition to the clinic, many KAT6A gatherings have taken place over the last few months. One of the best things about traveling is meeting up with other KAT6A families.

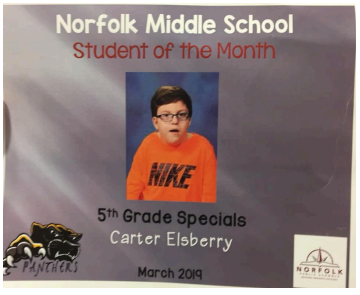
Sam stops by to see Holden in North Carolina on his way to Florida.

In Australia, Ginger's grandfather, Peter Harper, organized the 2nd annual **Omloop de Jindera** bike ride to raise funds for KAT6A. The ride was another success and raised AU$545.



In April, Natacha Esber attended the World Orphan Drug Congress in Maryland, USA to advocate for KAT6A. Thanks Natacha!

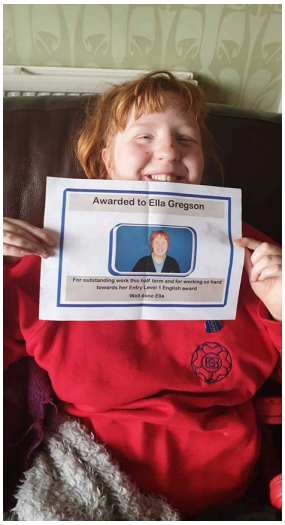
Anna, age 4, is making many new speech sounds and is really trying to talk! She can even say her own name. Way to go Anna!



Several of our children were honored in recent months for their efforts in school.

Congratulations to Carter Elsberry (left) who was selected as the student of the month at Norfolk Middle School this past March. Well done, Carter!

Ella (right) has worked hard in her English classes this semester and has received an award of achievement for her efforts towards English Level 1. Great job, Ella!



WHO WE ARE

Board of Directors

Karen Ginsburg

Jordan Muller

Emile Najm

Shelby Rau

Aimee Reitzen

Meg Salisbury

Jessica Vogland

David A. Woodbury

Chairperson

Jordan Muller

Chief Executive Officer

Emile Najm

Secretary

David A. Woodbury

Treasurer

Matt Salisbury

Communications

Director: Brittany Green

Family Services

Director: Tresh Crosby

Newsletter: Aimee Reitzen

Fundraising

Director: Jessica Vogland

Australia Region: Meg Salisbury

Merchandising: Shelby Rau

Science and Research

Director: Natacha Esber, MD

Registry Coordinator: Amy Young

Registry Support: Karen Ginsburg, Jordan Muller, Emile Najm, Andrew Rankin, David Woodbury

Social Media

Facebook: Aimee Reitzen,

Tresh Crosby

Instagram: Meg Salisbury, Kara Peschel

Twitter: Stephanie and Jake Langi

YouTube: Jessica Levesque

KAT6A Medical Providers

Austria

Dr. Sara Baumgartner, A.Univ.-Prof. Dr. Daniela Karall, IBCLC Medical University of Innsbruck, Clinic for Pediatrics/Inherited Metabolic Disorders, Innsbruck, Austria

France

Alain Verloes, MD, PhD, Chief of the Department of Medical Genetics, "CRMR Anomalies Développement & Syndromes Malformatifs et Déficiences Intellectuelles de causes rares", Robert Debré Hospital, Paris, France

Iceland

Hans Tomas Bjornsson, MD, PhD, starting June 2018

International

Richard Kelley, MD, PhD, Former Director, Clinical Mass Spectrometry, Laboratory, Kennedy Krieger Institute Associate Professor, Department of Pediatrics, Johns Hopkins University School of Medicine, Baltimore, MD

United States of America

Valerie A. Arboleda MD, PhD, Founder of the Arboleda Lab at UCLA, Department of Pathology and Laboratory Medicine, David Geffen School of Medicine, UCLA, Los Angeles, California. Dr. Arboleda will arrange referrals to clinical genetic specialists at UCLA.

Hans Tomas Bjornsson, MD, PhD, Director of the Epigenetics and Chromatin Clinic at John Hopkins University, Baltimore, MD

Jill Fahrner, MD, PhD., Assistant Residency Program Director at Johns Hopkins Genetic Medicine Residency Program and Assistant Professor of Pediatrics at Johns Hopkins Hospital, Baltimore, MD

Jacqueline Harris, MD, MS, Assistant Professor of Neurology and Pediatrics Director, Center for Tuberous Sclerosis and Related Disorders, Kennedy Krieger Institute, Johns Hopkins Medical Institution, Baltimore, MD

Kenneth N. Rosenbaum, MD, Founder of the Division of Genetics and Metabolism, Rare Disease Institute, at the Children's National Medical Center, Washington, DC