KAT6A News

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The 2020 KAT6A Clinic will take place on March 28, 2020!

For the third consecutive year, KAT6A families, international researchers and medical doctors will come together at the Kennedy Krieger Institute in Baltimore, Maryland, United States.

For the first time, we will have a Pre-Clinic day on March 27, 2020 and a social events schedule.

Online registration will open in January.

Read more on Page 5.

Inside This Issue

KAT6A Walk to Find a Way 2019

On September 14, 2019, the KAT6A Foundation hosted the 2nd annual Walk to Find a Way.

Thank you to all those who donated and participated. This year we raised over **$40,000,** making this the most successful fundraising event in our history!

Five official walks took place across the United States and the sun was shining in each city. We came together to walk in unity, raise awareness and funds for KAT6A research. In the process new friends were made and lots of love was received.

The largest events took place in Wantagh, NY and in Rowayton, CT. Each walk had approximately 100 participants. In Wantagh, three KAT6A families united for the first time making for an emotional day. In Rowayton, family and friends gathered for the second annual *Walk for Jack*. Amy Young said, "we were truly blown away by the amount of people that showed up to walk with us to help spread awareness of this rare disease. Overall, it was a wonderful event, and we continue to feel touched by the love and support everyone provides us during this incredible and often challenging journey with Jack."

In Woodbury, Minnesota, three KAT6A Families gathered with 50 additional walkers at a local park. In Boone, North Carolina, 30 walkers showed their support, and in Farmington, Arkansas dozens came out. The Crosby family held their own virtual walk in South Carolina to show their KAT6A love.

Walkers enjoy a sunny day at Cedar Creek Pak in New York.

Rachel and Emma Zielniuk at the walk in Arkansas.

The Vogland, Boles and Mrosla families gather at the walk in Minnesota.

The Young family raised over $28,000 at their Walk for Jack in CT, making them the highest fundraisers!

Chris and Holden Green pose for a photo.

The Green family came together in Boone, NC

We walk to find a way….

to get answers to the many questions about how KAT6A syndrome progresses.
to treatments that can alleviate our kids' health issues and developmental struggles.
to better inform medical doctors about KAT6A gene mutations.
for more research opportunities so all of this can be accomplished.
to bring families together each year at our KAT6A clinic.
We walk to find a way to a brighter future.

Above: John and Mary Najm pose for Instagram. Below: KAT6A moms: Natcaha Esber, June Sayers and Aimee Reitzen are all smiles.

Kevin Young discusses the goals and achievements of the KAT6A Foundation and the usage of donation money.

KAT6A Foundation at the 2019 NORD Summit

Dr. Esber, John Najm and Paul Najm stand in front of a giant visual featuring a photograph of them from the 2018 Summit.

On October 20, 2019, Dr. Natacha Esber represented the KAT6A Foundation at the "Member Organization Leaders" meeting in Washington DC. The meeting gave management tips as well as an overview of the NORD initiatives to help move rare disease research forward.

The 2019 Rare Diseases and Orphan Products Breakthrough Summit kicked off on Monday, October 21st with a greeting from NORD's President and CEO, Peter L. Saltonstall. Over two days, multiple keynote speakers took the stage at the Marriott Wardman Park Hotel in Washington, DC including Ned Sharpless, MD, FDA Acting Director and Alex Azar, Health and Human Services Secretary. "The Time is Now" was the theme of all the speeches and discussions between more than 900 participants across the rare disease community.

Dr. Esber, the director of science and research at the KAT6A Foundation, presented a Lunch & Learn Session entitled "Our KAT6A Journey, From 1 Patient to an International Organization".

John and Paul Najm, volunteers at the KAT6A Foundation, presented 2 posters: "My Journey from Autism to KAT6A" written by Peter Najm and "KAT6A Syndrome Explained by a Sibling" written by Paul and John Najm.

John and Paul Najm stand in front of their posters.

Amy C. Young attended the NORD Registry Meeting on October 23rd. Our registry launched in January 2019, and since then the team at NORD has been extremely helpful in navigating the completion of surveys and promoting participation. One of the most valuable takeaways Amy had from the meeting was being able to speak with other rare disease foundations' board members about their experiences and receiving useful feedback on how to make our KAT6A registry more successful going forward.

RESEARCH OPPORTUNITIES

KAT6A Patient 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.

One of the most important purposes of the KAT6A Patient Registry is to bring the KAT6A Syndrome community together and collect data which could be used to create therapeutics and improve the quality of life for patients. Some other goals of the KAT6A Patient Registry are to:

- Conduct a prospectively-planned natural history study that will result in the most comprehensive understanding of KAT6A Syndrome and its progression over time.
- Characterize and describe the KAT6A Syndrome population as a whole.
- Assist the KAT6A Syndrome community with the development of recommendations for standards of care.
- Assist researchers studying the pathophysiology of KAT6A Syndrome.
- Assist researchers studying interventional outcomes.
- Support the design of clinical trials for new treatments.

GET INVOLVED

What you want handy when completing the surveys:

- recent weight and height
- names of any publications the patient has been in
- genetic diagnosis (you have the option to upload a document or simply write out the diagnosis)
- medications & supplements (amount, usage, purpose)
- medical issues at birth
- any MRI or EEG results
- heart conditions
- digestive issues
- respiratory issues
- kidney and/or bladder problems
- hearing problems
- any allergies or immune system issues
- sleep disorders
-hospitalizations (when, length of stay, for what purpose)
- current therapy programs

Visit **http://www.kat6a.org/kat6a-registry/** to begin today!

For assistance email **kat6aregistry@gmail.com.**

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

KAT6A Clinic 2020

March 27, 2020: Pre-Clinic Day

Dr. Fahrner and Dr. Harris at John Hopkins kindly blocked off appointments exclusively for KAT6A families on this day. Schedule appointments ASAP.

Angela Morgan is coming from Australia and will meet with families consented to participate in the international speech study on KAT6A/B. Meg Salisbury of the KAT6A Foundation will assist in scheduling these appointments.

There will be a family friendly social gathering at the Baltimore Aquarium and in the evening dinner with KAT6A Researchers and Families.

March 28, 2020: Clinic Day

Kennedy Krieger Institute, 801 N. Broadway Baltimore, MD 21205 United States at Turner Auditorium

Tentative Schedule:

8:00 am: Welcome and light breakfast provided by KAT6A Foundation

9:00 am: Medical and Research Speakers: Dr. Valerie Arboleda, Dr. Jill Fahrner, Dr. Jacqueline Harris, Dr. Chaney Hollis, Dr. Richard Kelley, and Angela Morgan - SLP, Dr. Xiang-Jiao Yang, and a speaker from NORD.

Lunch- Provided by KAT6A Foundation

Afternoon WORKSHOPS: Valuable therapies, Financial Matters, KAT6A Foundation volunteers Q and A

Socializing with KAT6A Families, researchers and doctors

4:00 PM: Clinic ends

Dinner at restaurant located in Baltimore's Inner Harbor. Stay tuned for exact location and time.

KAT6A Clinic 2020 Registration

This year we will have a Registration Page for the event to help in planning and implementing a great clinic. Registration will open in JANUARY. It will be listed on **www.kat6a.org** and in our Facebook Support Group.

The KAT6A Clinic 2020 is a FREE event, sponsored by the KAT6A Foundation.

Please make sure you register for each member of your party because Kennedy Krieger Institute has a strict security policy and requires a names list in order for individuals to attend the event.

Add-on any OPTIONAL activities you would like to be involved in that weekend so that we can plan accordingly.

Add-ons:

- Social gathering at Aquarium on March 27th.
- Dinner Friday, March 27th TBD
- Dinner Saturday, March 28th TBD

Additional Information:

Children's room will be available, and childcare provided by volunteers, who are friends and family of KAT6A foundation members.

KAT6A merchandise will be available for purchase. Cash and checks only will be accepted.

****Check back often to our KAT6A support group on Facebook for clinic updates.**

In the Press

Photo of Jane and Carter taken by Margaret Burg

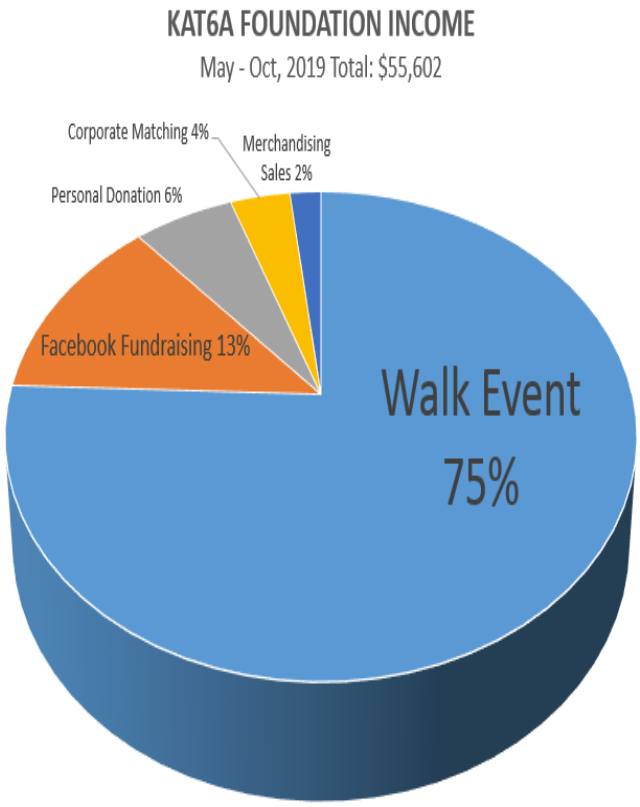
In September, Jane Ellul received a makeover by Carel Khairo from Beauty with Purpose. Beauty with Purpose has a mission to champion mothers who look after children with special needs by making them feel and look beautiful both inside and out. This was a special day for Jane to be pampered but also provided an opportunity to speak about Carter and KAT6A Syndrome. After hair and makeup, Carter and Jane were photographed by Margaret Burg in a natural and relaxed atmosphere. Carter is one of seven known cases in Australia. Jane says, "What Carel is doing is amazing. She is giving a voice to people to share and provide information to the greater community to help build more awareness. Thank you Carel, we need more people like you and your family in the world." Watch Jane advocating for Carter in this YouTube video. https://www.youtube.com/watch?v=LIFZ-yIMrKI&feature=youtu.be&fbclid=IwAR1IluQZDR1E1UlbdoY4NSJMTU5JqBSF--NQU87PrOt2UVpPWSnMgpjt0zE

In Perth, Australia, the Moura family have been advocating for Franki since he was born. After being diagnosed with KAT6A, they chose to undertake intensive therapy through Saba Rose Button Foundation's Rehab Me initiative. Franki's gains have been inspiring. Thanks to physiotherapy, Franki has recently begun walking. Read more about his journey at https://leahroberts.com/rehabme-intensive-collective-bringing-hope-to-perth-families/?fbclid=IwAR3XAZw89sW3QDpIEh0MEQbaNvU5xO9odNySP8q1GC4hs8dAC4inu-iEoFE.

Vera Moura, mother to Franki, was featured on a local television program called "Today Tonight". This was a wonderful opportunity for her to bring awareness to KAT6A. Watch her and Franki in action! https://www.facebook.com/TodayTonight/videos/780015905749333/UzpfSTUzNzE1ODg3NjoxMDE1Njg1MDk5NDc4Mzg3Nw/

In Warwickshire, UK, Daniel was able to meet his heroes, local police officers. Officers picked up Daniel and his parents from their home and took them to Bedworth Station for a tour. A film featuring Daniel's day was made by PC1942 to show important it is to engage with residents and communities. Also, helping to raise awareness for KAT6A syndrome. https://www.youtube.com/watch?v=2M5lJwYA8Js&feature=share&fbclid=IwAR0DARnC5WPL1VmsX5GhLTkyZV1eI_rrjNZ2LLkISHy3Wz2gzCKtp-dWR20&app=desktop

FUNDRAISING



Facebook Earnings News

A special thanks to the following individuals for showing support in 2019 via **Facebook fundraisers**:

Marc Monso Cairol, Amanda Campbell, Nord Chetbi, Tina Davis, Gladielis Diaz, Audrey Domsten, Miri Duston, Veronica Lices Fernandez, Sonia Gates, Matt Goes, Andrea Doner Goes, Olya Good, Britney Green, Rui Grosa, Kimberly Harvey Chase, Natacha Esber, Noe Sanchez Espinola, Christian Hor Vath, Simone Kabelitz, Adrian Khoo, Jacqui Mason, Tony McKinlay, Jordi Monso, Rick Perry, Patricia Richmond, Francesca Rivolti, Jeanne Syverson, Ashley Young Trowbridge, and Niki Watermolen.

In 2019, we have received over 400 DONATIONS on Facebook totaling over $12,000 USD!

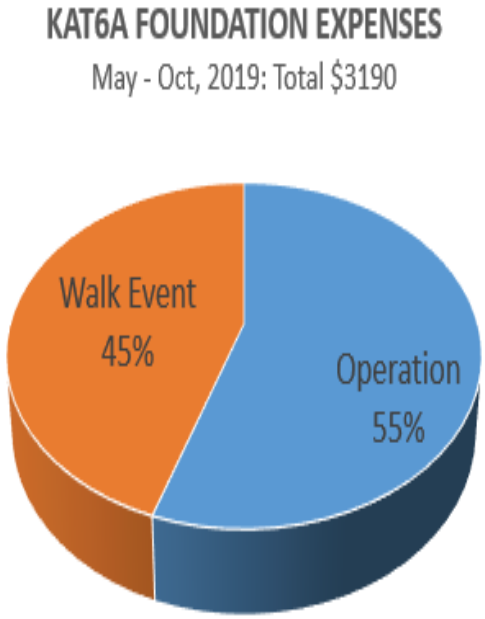
Ways to Support KAT6A Fundraising:

- Hold a Birthday Fundraiser on Facebook
- Shop using **https://www.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, or enquire about a matching program.
- Spread awareness on social media
- Join the **KAT6A Foundation Fundraising and Awareness Group** on Facebook.

#GIVING TUESDAY™

Share our KAT6A Foundation Giving Tuesday Fundraiser on December 3, 2019.

https://www.facebook.com/donate/511564252904356/



EVENTS

MARK YOUR CALENDAR:

Giving Tuesday

December 3, 2019

Giving Tuesday is the biggest single day for fundraising each year for nonprofit organizations around the world. Donations are matched dollar for dollar on a first-come, first-served basis until $7,000,000 USD in eligible donations made on **Facebook**. The Facebook donation match begins at 8:00 am Eastern Time. The match was met within seconds last year, so get your donations in early!

Rare Disease Day

February 29, 2020

Wear your stripes and KAT6A clothing on this day to raise awareness for our very rare community. Last year we had several families advocate for KAT6A in their communities, schools and even in the press. Be sure to look for our specialized photo frame on Facebook and use it on your profile picture.

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.

230 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, fundraising, or helping raise awareness, 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.

Where Does It Hurt?

by Emily Hoffines

Somewhere between winter and spring, when the days were warming up, but snowflakes still fell at night, I found myself sitting in the living room with my family. Each person was doing their own thing, one or two coloring at the table, one playing with toys, one reading, and of course Ruby was doing what she does best: a little bit of everything. From the corner of my eye I saw her tumble backwards and into a milk crate. It was sudden and caught me off guard, and I let out a chuckle because it had slap-stick comedic timing, but it took me a moment to realize that something was wrong. Ruby's arm was caught behind her, at a slightly odd angle, and she was crying so hard as to not make a sound. I yelled for Daddypants, who was closest, to help her up quickly, and he brought her to the couch, so we could check her out. She held her little arm limp at her side, cried silently, and buried herself into our arms. After peeling her shirt off to get a better look, we started asking,

"where does it hurt, Ruby? Here? Here? Here?"

We gently prodded different parts of her arm, from shoulder to elbow to wrist, but didn't get any reaction strong enough to shed light on where her pain actually was. There was redness at her shoulder where it hit the crate, redness at her elbow where she refused to bend it, and redness and swelling at her wrist. Ruby had previously had nursemaid's elbow, and we wondered if this could be the case again. However, reducing it would mean rotating her wrist so that her hand faced upward, and even the slightest turn of her wrist produced screams of pain. After applying ice and giving her lots of snuggles, we agreed that Ruby needed to be seen at the ER.

Ruby hates doctors. Going into the emergency room sparked a new cry from her. She was terrified and didn't understand what was going on. After getting checked in, we were called back for vitals. Before letting anyone get close to her, however, I explained who Ruby is, her fears, her history of having vitals taken, and her history of nursemaid's elbow. I also requested sedation. This is an awkward thing to request; “Hi, can you please sedate my child so she's easier to handle?” The woman who checked us in listened whole-heartedly and skipped the majority of the usual vitals and got us a room quickly. Then the doctor came in and stood back, asking me to be his hands and do the physical part of the exam while he watched and asked questions. "Where does it hurt?" Based on what he saw, he wanted to order x-rays. X-RAYS. Thankfully, they listened well, and brought in a nasal spray to sedate her enough to calm her for the x-rays. Unfortunately, they didn't know Ruby's strength, and I had to use all of my weight to wrestle Ruby and hold her as still as possible for the pictures and let me tell you I was sore the next day!

The films came back normal, but the doctor let me know that it's extremely common in children to be unable to see a break until it has already begun healing, so he recommended coming back for more x-rays in a week and putting her in a splint in the meantime. MORE x-rays. A SPLINT. Ruby was sedated again (nasal sedation wears off very quickly) to have her arm splinted, and we wrestled again (they doubled the dose this time) so the nurses could wrap her up. It was awful. I will tell you that getting the follow-up x-rays made getting the first set look like the easiest thing ever (they wouldn't sedate), but her arm wasn't broken! She suffered a bad sprain, and simply needed time to heal. And that week in a splint was enough to make me want to wrap her in bubble packaging.

Communicating pain is such a challenging obstacle for a child who is unable to speak, but more so for a child who is unable to understand the situation, the pain, and focus enough to try to get the message across. We have worked so hard with Ruby to use the sign for 'hurt', and sometimes if it's a small injury, or an obvious injury, we can "talk" through it. Scrape the knee? Kiss better, then sign that she hurt her knee. Bumped her head? Same thing. Stomach ache or headache? We'll have absolutely no idea.

I hope that someday, with continued practice and more experience, Ruby will be able to tell us not only when she is hurt, but also where the pain is. Until then it'll be twenty questions, lots of pointing, and hoping that we can get it figured out. I also hope that someday, someone will invent an x-ray machine that can capture films without scaring little kids so badly...I can wish.

International Affiliates

In Europe, there are now three independent organizations working to support families affected by KAT6A syndrome in their local countries. These associations are in Austria, Belgium and Spain. We look forward to building strong partnerships while working together to drive KAT6A research and awareness.

Contact Information:

Austria **https://www.instagram.com/kat6a_foundation_austria/**

Belgium **https://kat6abelgium.squarespace.com/**

Spain **https://www.facebook.com/Asociaci%C3%B3n-KAT6A-y-amigos-508123676595388/**

KAT6A Foundation Austria- MYKI Award Winner 2019

Sports Day in Tremp, Spain

In the Netherlands, Esmi Oostendorp started a blog about her KAT6A journey with son, Sep. She is hoping by spreading information about KAT6A in her Dutch native language that more awareness will be raised in her country. Check it out!
https://sepkat6a.jimdofree.com/

WHO WE ARE

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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.

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

Dr. Anne O'Donnell, MD PhD, Epichroma Clinic Boston Children's Hospital, Boston Massachusetts, USA

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