



# KAT6A News

Volume 2 | August 2018 | KAT6A Foundation, Inc.

## **KAT6A WALK TO FIND A WAY** **September 22, 2018**

This is a **VIRTUAL WALK** for all members of the KAT6A community and their supporters. On this day we will come together to raise awareness and critical funding for the KAT6A Foundation. The “mother walk” will be held in Raleigh, NC, USA, but there will be multiple walks occurring that same day around the globe. Wherever you are, we hope you will join us by starting a small walk in your neighborhood. Create a team or show your support by clicking on the link below. All donations will go directly to the KAT6A Foundation to support our mission to educate, raise awareness, identify new cases, fund KAT6A research, and ultimately find a treatment or cure. <https://www.classy.org/campaign/kat6a-walk-to-find-a-way/c189955>

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# FUNDRAISING and EVENTS

## Merchandising Success!

The 2nd KAT6A Bonfire T-shirt fundraiser raised \$878.47. We will be doing this fundraiser about twice a year. The graphic will change to keep it fresh and exciting.

The awareness bracelets have also profited more than \$200 in the last two months. We still have many adult and youth bracelets, so please check out the KAT6A Etsy shop to order. The bracelets are great to sell at any type of fundraiser you might be planning. Please contact Jessica Vogland for large orders. [jljohnson929@yahoo.com](mailto:jljohnson929@yahoo.com)

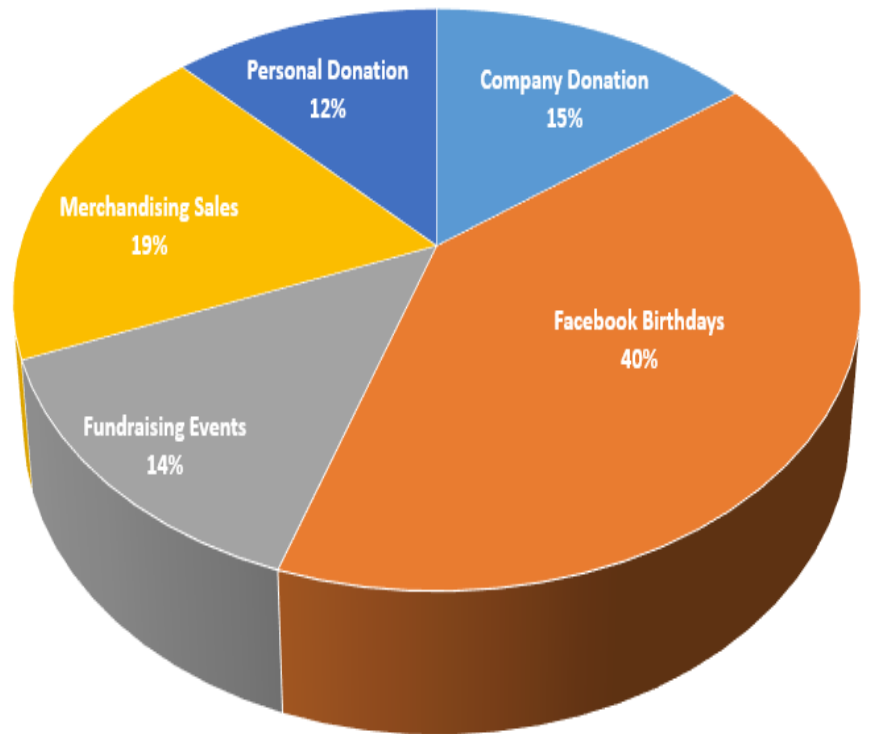
## Dalton Graham Run

July 21-22, The Geiger family raised awareness and financial support for KAT6A families at an event filled weekend to honor Dalton Graham. Dalton was the beloved uncle to our very own Savannah, who is living with KAT6A syndrome. The weekend included a picnic with basket raffle, motorcycle ride, and a fun barbecue lunch with family entertainment. Total raised = \$3033.83



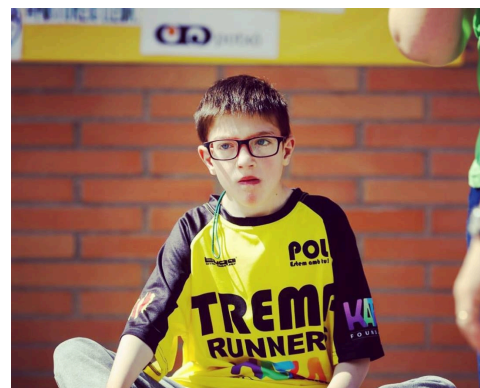
## KAT6A FOUNDATION INCOME - YEAR TO DATE

Jan - July, 2018 Total: \$20,699



## KAT6A & 10Q26 El Que Ens Fa Extraordinaris

On May 27<sup>th</sup>, a solidarity day was organized by the parent's association at Pol's school, son of Marc Monso Cairol, in Tremp, Spain. The fun-filled event raised awareness and research money for two rare disorders: 10Q26 and KAT6A syndromes. It was a great day in which many people took part. As a result, they raised 5112€ to be distributed between the Department of Rare Diseases of the Sant Joan de Déu Hospital of Barcelona and the KAT6A Foundation. Total raised = \$2854.00





## WAYS TO SUPPORT KAT6A FUNDRAISING:

- Hold a Birthday Fundraiser on Facebook
- Shop using smile.amazon.com
- Create a team for the KAT6A Walk to Find a Way
- Buy KAT6A bracelets on [ETSY](#)
- Purchase KAT6A awareness clothing
- 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.



*The Maas family helped their neighbors refuel and refresh with chips and water as they spread awareness for KAT6A and raised over \$70 for the foundation and for that, we say thank you!*



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

Aaron Albert, Dave Braska, Emmi Corpier, Tina Davis, Natacha Esber, Chrissy Elsberry, Emily Hoffines, Esther Kerner, Janis Neville, Kara Peschel, and Chloe Weddle.

## Save the Date!

### KAT6A Clinic 2019

February 9, 2019

Kennedy Krieger Institute

Baltimore, Maryland, USA

Details to follow

## VOLUNTEERS NEEDED!

We are looking for individuals to assist in collecting data for the **KAT6A registry** that our registry team is busy creating. Training will be provided. Please contact Emile Najm at [kat6a@yahoo.com](mailto:kat6a@yahoo.com).

We also need help spreading the word about KAT6A on social media. If you are interested in tweeting for the KAT6A Foundation on **Twitter**, or writing a blog post for the webpage, please contact Aimee Reitzen at [aimee4fh@hotmail.com](mailto:aimee4fh@hotmail.com).

## 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.

## 140 KNOWN CASES

[www.kat6a.org](http://www.kat6a.org)

Be sure to check out the KAT6A Moms and KAT6A Fathers videos on our [Youtube channel](#). Combined they have received over **5000 VIEWS** on Facebook! Cheers to all of you who submitted photos and shared the videos.

# RESEARCH

## Summary of Dr. Anne Voss's Conference: Experimental Treatments in KAT6A Mice

*Video available in the KAT6A Support Group*

On May 7th, Dr. Anne Voss at Walter and Eliza Hall Institute of Medical Research, Australia, held a teleconference about her work with mice. Over a dozen parents, as well as several of the medical professionals who are interested in KAT6A, joined the meeting, including Dr. Susan Hull, Dr. Andrew Rankin, and Dr. Richard Kelley.

Dr. Voss pointed out that she is a developmental geneticist, mostly interested in the brain.

Using a Powerpoint presentation, Dr. Voss spoke about modeling human embryonic development in mice, described the role of KAT6A in the packing and unpacking of the genetic material and showed a comparison between the KAT6A and KAT6B proteins.

For KAT6B, she showed photos of brain development defect in KAT6B deficient mice and demonstrated the heterogeneous mutations of the human KAT6B gene. She also showed the method to test potential treatments for KAT6B dysfunction in the mouse model.

For KAT6A, she showed pictures of the effect of the KAT6A heterozygous mutation on the mice brain and heart.

Her research plan for KAT6A and KAT6B is to model more patients' mutations in cells and in mice.

KAT6A mice experiments are already underway, based on Dr. Kelley's finding of amino acids imbalances in a subset of KAT6A patients. This study will compare wild type mice to heterozygous KAT6A mice. One group of mice (wild type and KAT6A) will be untreated, one group will be treated with Acetyl-Carnitine, one group with Valproic Acid and controls. The mice were treated for two weeks then the blood of these mice was collected and shipped to Baltimore, MD, USA for analysis by Dr. Kelley and his colleagues at John Hopkins.



Dr. Voss stated that experiments on mice are very expensive. She applied for funding in Australia and we'll know in November 2018 whether the

application for funding was successful.

She took many interesting questions:

-Among our KAT6A population, most of the boys don't talk, but a majority of the girls do talk somewhat. Dr. Voss pointed out that speech is a human specific skill, so the study of mice may not shed much light on this phenomenon.

-When asked how many KAT6A variants she plans to research in mice, Dr. Voss said that her first plan was to examine twelve, but the budget would need to be \$3 million Australian. She has scaled that back. Funding, she explained, needs to be planned for four years, because that is a normal time frame when doing cultures. Even by reducing the number of variants, she still has a budget around \$200,000 a year.

- It is good, she said, to have a whole organization, such as the KAT6A Foundation, asking for the research. Having clinicians and parents involved helps get an application funded. Our patient registry will help, too. Reviewers of applications ask for "significance." Dr. Voss is working on rare disorders. There is a bias in funding toward disorders that affect a high population. The true KAT6A population is probably much higher than we know at present. As whole-exome sequencing becomes more universally supported by insurance, more KAT6A cases will almost certainly be identified.

-Dr. Kelley added that all of his patients have rare disorders, but, from what he has learned so far, KAT6A seems to be one of the best disorders in which to show an important and under-recognized link between development and metabolism. He feels that a funding application should be well received, because it should be relevant to the treatment of all other histone acetylase disorders and many other syndromes.

## An Update on the Diagnosis and Treatment of KAT6A Mitochondrial Dysfunction

On July 9<sup>th</sup>, Dr. Kelley held a web conference for KAT6A families and medical providers. He started his talk by showing the KAT6A gene mutation effect on the intracellular level. With few slides he explained the mitochondrial dysfunction related to KAT6A by showing the interaction between the different enzymes in the nucleus, cytoplasmic space and mitochondrial space. He also explained the citric acid cycle (TCA cycle).

He presented data showing that autism is related to mitochondrial dysfunction as well. Then he showed a graph of plasma amino acid levels in KAT6A patients. Asparagine and Citrulline are elevated in multiple patients.

He then showed plasma amino acid levels of a patient treated with the mitochondrial cocktail who responded quickly and clearly due to the cocktail. After a year of starting the cocktail, the patient's amino acid levels were all within normal range. The patient's mother spoke about the clinical improvement that she noticed after starting Carnitine, such as improved feeding abilities, gross motor functioning, and alertness. Furthermore, her son's constipation resolved within weeks.

Dr. Kelley recommends the following mitochondrial treatment for KAT6A patients:

### KAT6A Mitochondrial Treatment

#### Prevention of Fasting Stress

Cornstarch 1 g/kg HS

#### Complex I Activation

Carnitine 50 mg/kg/d

Pantothenate 3 mg/kg/d

#### Antioxidant Protection

Coenzyme Q10 10 mg/kg/d

Vitamin E 15 IU/kg/d

Vitamin C 40 mg/kg/d

#### Amino Acid Supplements PRN

Methionine, Tyrosine 10-30 mg/kg/d

He also stated that citric acid supplementation is helpful for gastrointestinal motility in mitochondrial dysfunction after ruling out an anatomical abnormality.

*Visit the files section of our support group to view a full slide presentation of the conference call.*



We are pleased to inform you that NORD's Board of Directors has approved the KAT6A Foundation to become a NORD (National Organization for Rare Disorders) member organization.

We look forward to a long and rewarding collaborative relationship with NORD as we will work closely together to represent the needs of people with KAT6A gene mutations.

NORD reaches millions of physicians and medical professionals through medical publishing partnerships, online resources such as WebMD, and social media outlets. Therefore, NORD will be a vital tool for our foundation in increasing KAT6A awareness.

Additionally, NORD will assist us in building the first KAT6A patient registry. The data compiled in the registry will be a valuable source of information for researchers and members of our KAT6A community.





## Life on the Other Side

by Beth and David Woodbury

When parents have a baby or child with a serious diagnosis, there are many unknowns. One is the future—what will happen as my child grows up. Will his siblings resent all the attention I give him? Will he have friends? Will he have a place to belong? Will his life have meaning? Will I be able to meet his needs? Will that be enough? Are all our dreams of the future shattered?

Here is Sam's story.

Sam was born in August 1990 six weeks prematurely. Nothing was right, even from the start. He didn't breathe, he couldn't regulate his body temperature, and he couldn't eat. And things didn't get too much better very fast. He had bradycardia, he had strange breath-holding spells (or were they seizures?) when he would turn blue and pass out. He spit up a lot, he had terrible screaming spells, he had pneumonias, he wasn't moving normally, his developmental milestones weren't being met...he was blind. That was just the first four or five months. At eight months we found out he had an intestinal malrotation and needed surgery for that as well as a nissen fundoplication and g-tube placement. That was the first of many intestinal surgeries and hospitalizations. At age three there was another suspicion that was confirmed—he also had autism. We spent so much time at Maine Medical Center in Portland, Maine, that he and I virtually lived there. My husband and our older daughters, ages 11 and 14, shuttled back and forth to "visit" us. There were so many doctor and therapy appointments, so many evaluations and tests, procedures and surgeries that about four or five years passed in a blur. See, I don't even know how many years!

But gradually Sam was better intermittently, and we adjusted to our new life. He eventually sat up, crawled, and learned to walk with a walker. He did not learn to talk, and he still had a lot of medical problems and developmental delays. He had insomnia. It turned out that he wasn't blind in the ordinary sense, but he did have cortical visual impairment. It was pretty clear that he was never going to be very much like any other kid. He had very strange, maladaptive behaviors and a lot of screaming when things weren't the way he thought they should be (like, if the car turned right and he liked left turns). It was pretty clear he was not going to be very much like any other kid. There was a lot to be scared about.

He started "home" school at a few months of age — vision therapy, physical therapy, occupational therapy, developmental therapy, and then some kind of autism therapy. By age 2.5 he was off to special preschool in the mornings and the individual therapists came in the afternoons. Too much therapy!! But at age 4 we began a new kind of therapy, Applied Behavior Analysis — at school and at home. That was a great turning point. We finally had everyone on the same page, working as a team. And if they didn't want to get on board, they got out. We were learning too.

About the time Sam was turning five, our family made some decisions. It was pretty clear that I couldn't work outside the home because there was no daycare or provider for Sam. His doting sisters were now 19 and 16 and it was pretty clear that Sam was the center of the universe to all of us...was that the best thing for him? For us? We had the bright idea that we could put our newfound skills to work, give me a "job" so I wasn't so totally focused on Sam, and provide siblings to Sam nearer his age. A win-win!! The job? We became a treatment foster family. This means that we provided a home to kids with developmental or mental health issues.

Ho ho ho! and it wasn't even Christmas! First came a couple of sisters aged 11 and 12 — not exactly the little boys Sam's age that we had pictured, but they definitely became siblings, made our lives more lively, and kept us from over-focusing on Sam. They stayed with us until they were ready to spread their wings and fly. Next came another girl...this time only six years old, then a few years later a girl aged 11. Again, these kids were permanent siblings — Sam, and the two younger girls were each only a year apart in age. We had many others in and out temporarily while they were either working on returning to their birth families or moving on to a different situation, or just for regular respite.

So Sam was never lacking for siblings...especially sisters. It wasn't always easy, it wasn't always pretty, but it was our family. It was a hopping household for sure.

Meanwhile, Sam grew, and grew older. He had his intermittent emergency hospitalizations, usually related to his gastro-intestinal abnormalities. He had a few surgeries, lots of doctor appointments, lots of testing, some ups, some downs. When he was 10, we had to leave our hometown due to the paper industry going belly-up...from human resources in a very large paper company, David moved to human resources in a small hospital, and we moved 50 miles down the road. We decided it would be a good move and adjusted to new providers, a new school system and a new house. And good news! We were now 45 minutes closer to the nearest service town! Since we have always had to make that trip to Bangor at least once a week and sometimes daily, that was a big deal. And the ambulance could get to Portland 45 minutes quicker too...always look on the bright side of life!

And so time passed. Sam went to school — there were ups and downs. He developed an exaggerated startle reflex, hyperekplexia, which further compromised his ability to walk. We spent untold hours advocating at the state and local level for appropriate services. We had a few unpleasant experiences and we had a lot of pleasant experiences. He had some wonderful teachers and after-school staff, he had a few not-so-good ones. We traveled a lot: by planes, trains, and automobile and had a lot of amazing adventures with our crazy family. Sam never learned to walk independently or to talk, but he learned to get around and he figured out how to communicate in his own way. (He had to with all those sisters!) He made friends, oh boy, did he make friends! Time flew by, and then it was time to cross over into the dreaded "other side" ...adulthood.

And you know what?? It is great here!

Bored? Lonely? Stuck at home? Never!! Over the past few years Sam has truly found his place in life.

One advantage of living in a very rural area is that there are no "day programs" — Sam has one-on-one staff for 37.5 hours a week. They are busy — with Special Olympics, their self-advocacy group, friends, and community groups all over town. He has one special volunteer job, and participates in lots of other special projects, for instance, they are planning a special KAT6A fundraiser for spring, because Sam finally has a diagnosis, and a cause, and his friends want to help. Sam has more friends than I do!

He still lives at home with us because that is what we choose. And because we find it more difficult to travel now that Sam is a grown man, we have turned our property into a full-fledged redneck resort, just for Sam and our family and friends. If we can't go to them, we want them to want to come to us, and they do come! Three of his sisters left home, went out to explore the world, and then came home to Maine. Along the way they found and married three very special men, who love Sam almost as much as his sisters do. Another sister lives in a nearby town. There are children, dogs, babies, friends, and family underfoot constantly...and adventures to be had with all of them. Uncle Sam is a great favorite — who else's uncle can play with toys the way he can?? And Sam has lots of fun grown-up toys everyone wants to share!

He makes a difference to many and has a fulfilling, meaningful place in his family and community. His greatest gift is making people feel loved—his smile can light up a room and his hugs make troubles melt. He participates. He contributes. There isn't anyone who knows Sam who doesn't love Sam. What more could anyone want?

It is not the life I pictured when I married my handsome prince nearly 43 years ago. But it is a far cry from the life I feared and worried about twenty years ago. **Life is good here on the other side with Sam.**

## NOTEWORTHY ACHIEVEMENTS

We all know how difficult potty training a child can be, but potty training a child with KAT6A brings a unique set of challenges. I think we all would agree as to what an achievement this is. Way to go Parker! He just “pooped on the potty!”



Emma in New Jersey is also working hard to potty train and her family is celebrating her waking up dry.

Many of our children experience sensory issues and food intolerances making eating a difficult task. Victoria experienced ice cream for the first time.



Several in our community attended prom this year. Here is Ellie from England dressed for her big night out.

Little Korra is learning to blow kisses.

Many of our kiddos have learned to ride bikes this summer and their overall gross motor planning has improved. Bay began riding her y-trike and has made big strides in purposeful play. Honorable mentions to Claire who quickly moved from a trike to a bike, and Cullen who started riding a two-wheeler independently.



Will began picking up his food independently with a fork.

Franki from Australia has begun crawling after several intensive sessions of physical therapy.

Jack initiated his first steps with his walker this summer. He and his new baby sister



Ella from the United Kingdom is all smiles having been selected to be a school prefect for this upcoming school year. She gets to wear a red sweatshirt for her new title. Congratulations, Ella!

Sam earned 2 bronze medals in the Maine Special Olympics in the 10 and 25 meter assisted walks, as well as two 4th place finishes in bocce and softball throw. Congratulations to our Olympian!

Many of our children attended camp this summer and are taking leaps towards independence. Toby in Canada attended scouting camp.



Tommy in California attended summer camp and began walking independently. Tommy’s father says, “After 3 years with a walker Tommy tossed it aside for the freedom of independent walking and he has found his ability to choose his own activities, too! He also discovered a special needs trike at summer camp and has a natural ability to pedal and steer without being taught!” Way to go, Tommy!



## WHO WE ARE

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Tresh Crosby

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Aimee Reitzen

### 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



# KAT6A AWARENESS GALLERY



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