



Volume 3 | December 2018 | KAT6A Foundation, Inc

KAT6A CLINIC

February 9, 2019

9:00 am – 3:00 pm

Kennedy Krieger Institute

707 North Broadway Baltimore, MD 21205

This is the second annual KAT6A Clinic. It is a unique opportunity to learn from distinguished medical professionals and to meet other families coping with this rare disorder. The events speakers include: Dr. Jacqueline Harris, Dr. Richard Kelley, Dr. Anne Voss and Xiang-Jiao Yang, PhD.

Breakfast and lunch will be provided by the KAT6A Foundation, Inc. Childcare will also be available. RSVP on the events section of the [Facebook](#) page of KAT6A Foundation. Remember to wear your KAT6A clothing to the clinic.

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MESSAGE FROM THE KAT6A FOUNDATION

Dear KAT6A Community,

As 2018 comes to end, we reflect on the first full year of the KAT6A Foundation and look forward to 2019. We began as eight families with a common desire to advance KAT6A research so that we could discover a treatment that would improve the quality of life for our children. Although this remains a strong part of our mission, our motivation has vastly expanded. We proudly exist to support KAT6A families in any way we can.

We successfully united families and experts at our first KAT6A Clinic last February. This was a unique opportunity to educate our support group members on the rare disorder by experts in genetics. It was the first time that many parents were able to meet other KAT6A parents coping with similar challenges and to finally see other children with their child's diagnosis. In a room full of people who had never met before, there were no strangers. We look forward to bringing together even more families at our second KAT6A Clinic on February 9th.

It is a critical part of our mission to educate KAT6A families, teachers, therapists and doctors. Our website has become the key resource for families and medical professionals. The KAT6A Foundation provides the most current information regarding this rare syndrome. Our knowledge of KAT6A will expand after the launch of the registry in 2019 and as findings are revealed from our current research studies.

We strive to support individuals with KAT6A by raising awareness about this rare disorder and by spreading a message of acceptance. We opened our online store this year, so that KAT6A families can proudly let the world know who we are. Every time we wear our KAT6A clothing, we have the opportunity to educate others. On social media, we have many loyal followers and are looking to expand these numbers so that one day KAT6A will be largely known.

In 2018, our support group doubled in size as we connected to more families internationally. There are now over 165 known cases in 27 different countries. Today, when a family receives a KAT6A diagnosis they can readily find information, and we can begin offering them hope, guidance and support immediately. Nobody needs to navigate this rare diagnosis alone. We are proud of the strong bonds formed in our global family via social media outlets and technology; bonds that have encouraged local gatherings and friendships.

We are overwhelmed with gratitude for our donors. Thank you, 90,000 times, over. Each and every dollar will make an impact in the lives of KAT6A individuals as we invest in research, provide materials in multiple languages to inform KAT6A families, sponsor events to bring families together, and move forward with our mission to ensure that every KAT6A person will receive the medical and therapeutic treatments they need in order to reach their full potential. Many individuals supported us again and again through website donations, giving to one of our events, such as the inaugural KAT6A Walk to Find a Way, purchasing merchandise from our KAT6A shop or Bonfire campaigns, and on Facebook fundraisers. We want you to know that we recognize you and are humbled by your generosity and will rely on your continued support in 2019.

Finally, we want to thank all our volunteers who offer their invaluable time and resources in order for us to pursue our mission. If you are interested in joining us, there is a role for you to fill. We are eager to press forward in the new year and to continue growing as a foundation.

Happy holidays and a prosperous new year from all of us at the KAT6A Foundation!

FUNDRAISING and EVENTS

KAT6A Walk to Find a Way

On September 22, 2018, the inaugural KAT6A Walk to Find a Way took place. The main event was held in Raleigh, North Carolina. Satellite walks took place in Connecticut, New York, New Jersey, Minnesota, Missouri, Canada, Spain and Australia. It was a joy filled day in which KAT6A families and friends raised awareness and over **\$25,000** for the KAT6A Foundation. Thank you, Blake Hiott, for dreaming up this event, for orchestrating the Raleigh walk, providing a barbecue, silent auction, t-shirts for all walkers, and decals for families. Way to go Young family for your outstanding fundraising achievement in Connecticut.



The Braska family with Blake Hiott (right) pose for a photo at the check presentation. David Braska was the inspiration for this incredible event.



Shop for KAT6A

Our official **online shop** launched in October with the perseverance of Shelby Rau. This exclusive marketplace allows individuals the ability to purchase all items whenever they choose to do so. Our online marketplace includes clothing, water bottles, mugs, aprons, phone cases, clothing for babies, and even bandanas for your puppies.

We still have many adult and youth awareness bracelets, so please visit the KAT6A Etsy shop to order. The bracelets are great to sell at any type of fundraiser you might be planning. Contact Jessica Vogland for large orders at jljohnson929@yahoo.com.

#GIVING
TUESDAY™



A special thanks to the following individuals for showing support via **Facebook birthday and "Giving Tuesday" fundraisers**: Roxan Boles, Amanda Campbell, Tresh Crosby, Lindsey Geiger, Stephanie Langi, Lin Lucas, Emile Najm, Shelby Rau, Aimee Reitzen, Traci Starn, Ashley Trowbridge, Katie Ward, Casey Williams, Beth Woodbury

Total= \$5,769 raised this quarter

Get Involved!

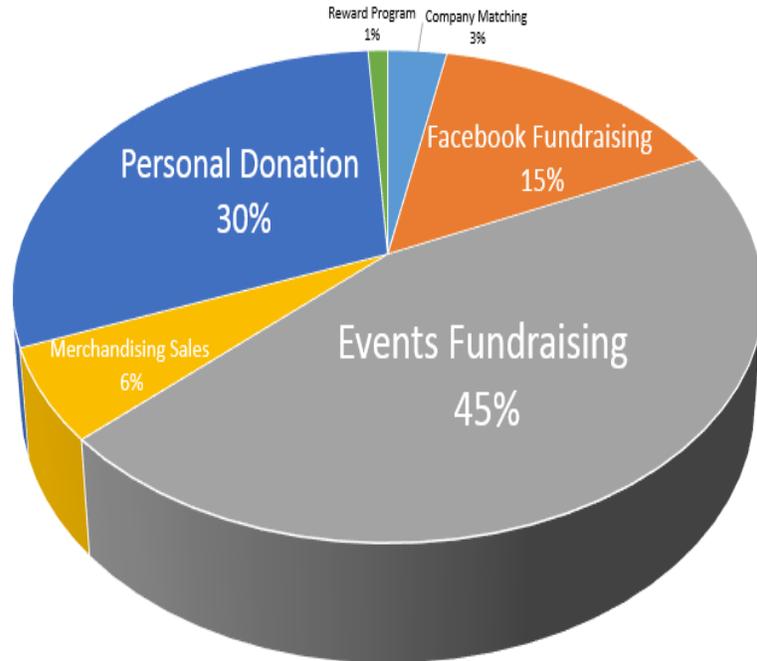
KAT6A FOUNDATION INCOME - YEAR TO DATE

Jan - Dec, 2018 Total: \$72,772

We are looking for people to join the **KAT6A Fundraising and Awareness committee**. If you are interested in joining us to raise funds and bring awareness to KAT6A we would love your help. Please contact Jessica Vogland at jljohnson929@yahoo.com. The first meeting will take place at the beginning of 2019.

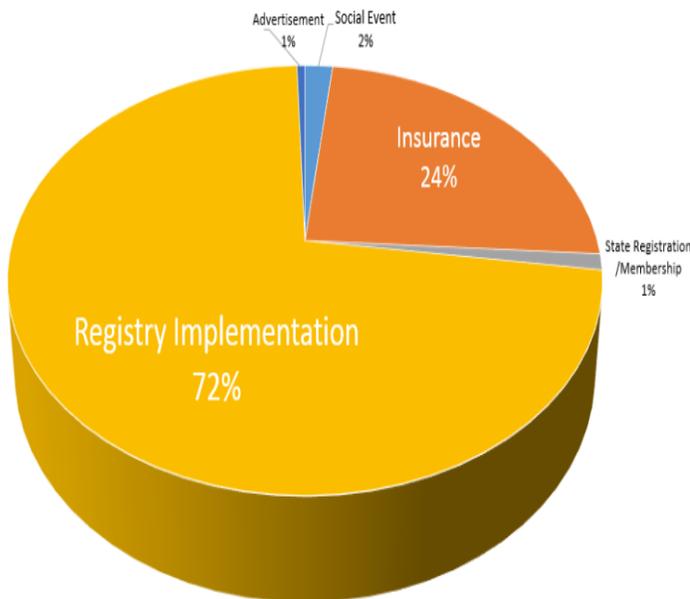
We need volunteers to assist in collecting data for the **KAT6A registry** that our registry team is busy creating. Training will be provided. Please contact Emile Najm at kat6a@yahoo.com to get involved.

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 support@kat6a.org.



KAT6A FOUNDATION EXPENSES - YEAR TO DATE

Jan - Dec, 2018 Total: \$ 6,917



Ways to Support KAT6A Fundraising:

- Hold a Birthday Fundraiser on Facebook
- Shop using smile.amazon.com
- Buy KAT6A bracelets on [ETSY](https://www.etsy.com)
- Purchase KAT6A awareness clothing and accessories from our [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.

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.

165 KNOWN CASES

Use this link for your next Facebook fundraiser.
<https://facebook.com/fund/kat6afoundation>
Remember, there are no fees collected on Facebook, so 100% of the donations made will go to our nonprofit.

RESEARCH

Summary of Dr. Valerie Arboleda's Conference Call: KAT6A Syndrome: Genotype-Phenotype Correlation

Video available in the KAT6A Support Group

Dr. Valerie Arboleda at UCLA held a webinar for KAT6A families on October 29 to give an overview of what she and her team have done and where her research is headed. She introduced the subject by reviewing the just-released article in *Genetics in Medicine* on the study of “76 patients with pathogenic KAT6A variants”. What is KAT6A? It is the gene that encodes lysine (K) acetyl-transferase protein. K is the short code for lysine, which is an amino acid. It's also the K in KAT6A. Proteins do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs.

Acetylation is important in two functions: chromatin packaging (how the DNA molecule is literally crammed into the nucleus of every cell in the body), and regulation of proteins (turning proteins on and off, how proteins move within the cells, and more).

There can be missense, protein-truncating, or splicing mutations of the KAT6A gene. Arboleda described each of these mutations and their effects. A missense mutation may not interrupt the cell's synthesis of a protein but changes how the protein is made and therefore what the protein can do. Truncating mutations are the vast majority of changes and cause the KAT6A genetic syndrome. A splicing mutation may be truncating but in a different way — a vast majority of them do result in a truncated protein molecule. It occurs when a protein is being built but the coding skips a number of nucleotides in the RNA.



Arboleda's team and Backus Lab enjoy some fall fun.

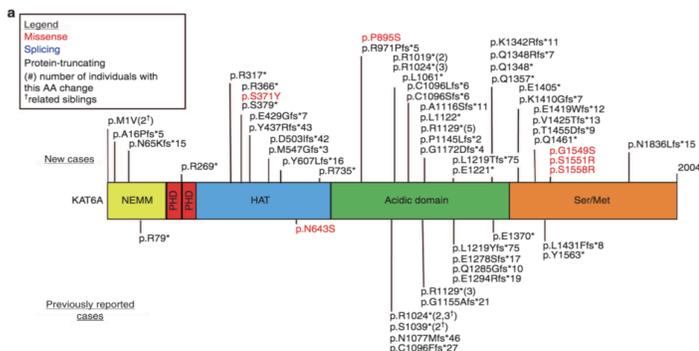
Arboleda's team was able to identify a number of these mutations with disease features. The most consistent features across KAT6A are a language deficit and some intellectual disability. They found that the social smile in an infant comes at the appropriate time. Walking independently is quite variable. They identified several common facial features, but these are not sufficient for diagnosis. Gastro-intestinal and mitochondrial problems are less common. A summary of clinical features can be found in Table 1 in the article along with clinical advice and guidelines in Table 2.

She discussed treatment versus cure. KAT6A is impossible to change across all cells in the body. A treatment might modulate certain symptoms. She believes we're a long way from identifying a single drug or group of drugs that might have an effect on the symptoms of KAT6A mutations.

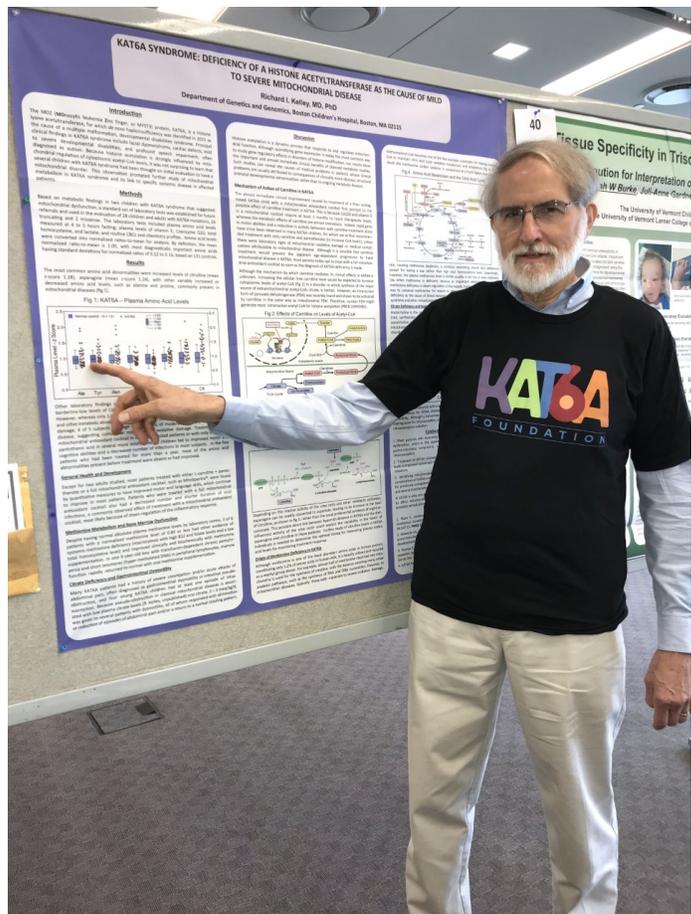
Her future work includes a new survey on treatment modalities and collecting saliva samples from all KAT6A families. Opportunities to participate in her research will be shared with members of the KAT6A support group as they become available.

KAT6A Research in Israel

Dr. Yehuda Assaraf of Technion Institute in Israel has selected four KAT6A subjects who generally represent the spectrum of presentations and will soon begin a multiplex analysis of KAT6A Function in cells derived from these individuals.



Dr. Kelley Poster Presentation in Alberta



Dr. Richard Kelley presented a poster at the 39th Annual David W. Smith Workshop on Malformations and Morphogenesis that was held in Alberta, Canada from August 24 till August 29th, 2018. The title of the poster is: KAT6A SYNDROME: DEFICIENCY OF A HISTONE ACETYLTRANSFERASE AS THE CAUSE OF MILD TO SEVERE MITOCHONDRIAL DISEASE. Here's a summary of the poster:

The MOZ (MONocytic leukaemia Zinc finger or MYST3) protein, KAT6A, is a histone lysine acetyltransferase, for which de novo haploinsufficiency was identified in 2015 as the cause of a multiple malformation, developmental disabilities syndrome. Because histone acetylation is strongly influenced by mitochondrial regulation of cytoplasmic acetyl-CoA levels, it was not surprising to learn that some children with KAT6A syndrome had been thought on initial genetic evaluation to

have a mitochondrial disorder. This observation prompted further study of mitochondrial metabolism in KAT6A syndrome and its link to clinical disease in patients.

After the referral of several children with KAT6A syndrome beginning in 2016, a standardized set of laboratory tests was established and used in the evaluation of 28 children and adults with KAT6A. These tests included plasma amino acid levels measured at 4 to 5 hours fasting; plasma levels of vitamin E, Coenzyme Q10, total homocysteine, and lactate; and routine CBCs and chemistry profiles.

The most common amino acid abnormalities were increased levels of citrulline (16/28; mean metabolic abnormalities diagnostic of mitochondrial disease, 4 of 5 subjects over age 10 had moderately severe mitochondrial disease, suggesting cumulative, age-related oxidative damage. Treatment with a mitochondrial antioxidant cocktail in severely affected patients or only carnitine + pantothenic acid (to augment complex I activity) in several more mildly affected patients led to improved motor and cognitive abilities and a decreased number of infections in most treated subjects. In addition, despite having normal absolute plasma methionine levels, 3 of 6 subjects with low normalized methionine levels had other evidence of systemic methionine deficiency (anemia, low total homocysteine level, high MCV).

Both histone acetylation and methylation are dynamic processes that respond to and affect mitochondrial function, among other aspects of cellular metabolism. Although quantifying gene expression is a standard way to study gene regulatory effects in disorders of histone modification, the results reported here show the utility, including immediate clinical benefits, of planned metabolic studies to find the causes of clinical disease in syndromes whose clinical problems are often attributed to their associated prenatal developmental abnormalities.

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

The KAT6A Foundation and the National Organization for Rare Disorders (NORD) are working to launch the largest-ever study to research KAT6A Gene Mutation that causes KAT6A Syndrome.

The 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 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 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.

On Wednesday, October 17 Jordan Muller and Amy Young attended the NORD IAMRARE™ Registry Community Meeting. Twenty-four representatives from sixteen member organizations joined the NORD Research Team in Washington, DC, following NORD's Rare Diseases & Orphan Products Breakthrough Summit.

Amy Young stated, "Although I learned a lot about how NORD will help our KAT6A Foundation once our registry is up and running, I found that the most beneficial part of the conference was having the opportunity to speak with other representatives who were just like I am- a concerned parent who wants to learn more about their child's rare disease!"

Some of these member organizations have had their registries live for 3 years, and it was extremely helpful to be able to ask them questions about what worked and didn't work when it came to spreading awareness of the registry and encouraging hesitant families to complete it. I am excited to continue working with NORD to make our registry live in the next few months. I walked away from the Registry Community Meeting feeling comfort in the fact that NORD exists in order to best help support Jack and other children with rare diseases."

Be sure to read the new report on [KAT6A Syndrome](#) located in NORD's Rare Disease Database. Look for our registry to go live in 2019.

Rare Diseases & Orphan Products Breakthrough Summit

Dr. Susan Hull, Dr. Natacha Esber and Paul Najm attended the "Rare Diseases & Orphan Products Breakthrough Summit" of NORD on October 15-16, 2018 in Washington, DC. The Summit theme was "A New Era of Patient-Focused Innovation" and it featured key speakers from the FDA.

The KAT6A foundation presented 2 posters at the Summit: "KAT6A Syndrome: Deficiency of a histone acetyltransferase as the cause of mild to severe mitochondrial disease" by Dr. Richard Kelley and "Life-changing effect of L-Carnitine in a patient with KAT6A syndrome" by Dr. Natacha Esber and Paul Najm.



Paul Najm, son of Natacha Esber, stands by their poster at the NORD summit.



Holidays with Holden

by Brittany Green

Christmas has always been a big deal in our house. All of us, kids and parents, enjoy the feeling of Christmas. We especially love the glittery, sparkly decorations that come along with it. We always have lots of garland, lights, and decorations in our house—you know—all the sparkly fun things kids love!

When you have a special needs child, those things can be either wonderful or horrific. For Holden, who is three and a half years old, it depends on his mood of the day. This year, when we pulled the Christmas tree out of the bag for the first time, we thought poor Holden was going to have a heart attack! A big, green, prickly, scary thing in the house! Holden must have thought it was a big green monster. But we got it set up with all the decorations and lights and he began to be curious about it. Pretty soon he was pulling ornaments off the tree. As the mom of three boys, I will be the first to tell you that the last thing you want is a child pulling ornaments and decorations off your beautiful tree. But when Holden began to do that instead of showing fear or aggravation I was happy! We were all relieved he was no longer scared of the tree and was enjoying having it in the house. We let him explore and figure out that the Christmas tree was harmless and was not going to hurt him.

Like many kids with special needs, Holden does not do well with change, and he has become much more observant of his surroundings over the last year. So many different things can upset him and cause him to be very unhappy. Now that it is the Christmas season, our house is not looking the same as it always does. The first few days with the decorations, he was really confused and upset about the changes. With a lot of patience and some time, Holden has become used to all the new thing in the house and is actually starting to enjoy some of them.

Since we never know what simple thing might be the one thing that he just doesn't agree with, we were worried about what his reaction to Santa Claus would be. Surprisingly, Holden was perfectly fine seeing Santa and sitting on his lap! His older brothers were sitting next to him, so that made him feel safe and secure.

We hope everyone has a blessed Christmas season.

Love,
The Green Family

Jack Makes a Splash

by Amy Young

It has been an exciting few months in our household. We welcomed our daughter, Emma, on July 1st, 2018. Jack has been a wonderful big brother thus far. He is beginning to notice Emma more and more each day; there are even times when he seems annoyed by his little sister (as a typical three-year-old might)!

Even though life has been quite hectic since our newest addition arrived, we have made sure to continue with all of Jack's weekly therapies. One therapy in particular that has made an impact is Aquatic Therapy. Jack works with a PT in the water one on one every Wednesday morning for 45 minutes. Not only does Jack truly enjoy swimming, but the benefits are clearly evident. Each session provides Jack with the opportunity to improve core strength, mobility, balance reactions and range of motion. He is happy and motivated to move in the water. The therapeutic properties of the water allow Jack to move more freely and independently. He tolerates handling, stretching, and active exercises, in conjunction with lots of sensory input. Jack often smiles throughout his sessions and happily kicks and splashes his way around the pool. It goes without saying that physical therapy is so important for our KAT6A kids, but we can honestly say that Jack would not be making all the progress that he is without his weekly time in the water!



David Woodbury: Personal Story from the New Era of Patient Advocacy

The KAT6A Foundation is thrilled to announce that David and Sam Woodbury's story for patient advocacy was selected as one of only 35 stories chosen to feature on NORD's YouTube channel as part of their campaign "35 Years and Growing: Personal Stories from the New Era of Patient Advocacy." David and his wife have dedicated their lives to advocating for Sam and the rare disease community since they learned of Sam's medical challenges. We are so proud this incredible family. Enjoy viewing their [video](#).



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NOTEWORTHY ACHIEVEMENTS

Difficulties in expressive language seem to affect most of our KAT6A community to varying degrees. Facial expression, assistive technology, sign language, and for some even vocalizations are but a sampling of the tools our family members utilize to communicate. Being understood is no small accomplishment, and our friend, Huddy, has made tremendous leaps in becoming bi-lingual recently using American Sign Language. Well done, Huddy!



Hayley has participated in therapeutic riding since she was three years old and in 2017, decided to take a break from track and field Special Olympics events and enter the equestrian events instead. This year she earned not one, but two silver medals! Congratulations, Hayley! We are so proud of you.

And speaking of track and field, congratulations to Ryder for earning three personal best times in 100 m running, long jump, and discs. His mother says that even though he comes in last in every event, “you can’t wipe the smile from his face.” Celebrating you, Ryder, and what you can do! Well done.



Our children are so unique and on their own individual developmental clocks. Let’s celebrate with Eimy as she is now able to hold her head up long enough to watch some TV. What a HUGE accomplishment, Eimy!

And how about this milestone! Madison has learned to walk like her big sister. So much love and admiration here. Bravo, Madison!



Isaiah has learned to turn and stop on skis this year. That’s certainly more than most adults can do! Maybe you can offer classes to a few of us. Congratulations!



And perhaps the most remarkable “notable achievement” of all is the coming together of families from all over the world who pour their lives into their children and other families of the KAT6A Foundation. To the left are Will’s parents from the USA meeting Ginger’s parents from Australia for the very first time. Remarkable!



WHO WE ARE

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MD

Alain Verloes, MD, PhD

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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

Would you like to be featured in our next newsletter?

Contact support@kat6a.org and we'll share your story in our next installment of KAT6A News.

