If your loved one was diagnosed with a KAT6A gene mutation, we are here to inform, support and aid you in coping with the daily challenges associated with this very rare syndrome.

What does a KAT6A gene mutation diagnosis mean?

KAT6A is a rare syndrome resulting from a mutation in the KAT6A gene. Through genetic research, we have learned that the KAT6A gene makes the KAT6A protein. The KAT6A protein is involved in controlling the production of proteins from other genes. Therefore, when there is a change on the KAT6A gene, problems can occur in various parts of the body. Currently, scientists do not know all of the functions of the KAT6A gene so our knowledge will increase as research advances.

Through 2017, about 100 people have been diagnosed with KAT6A from around the world. Each person with KAT6A syndrome has a different mutation along the KAT6A gene, which leads to a range in symptoms and features. 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.

It is important to note that many parents describe their children as happy and healthy despite global developmental delays. The diagnosis is an important step in helping your loved one but it does not change the person that they are.

Please visit the official website of the KAT6A Foundation. We hope you will access the About KAT6A page to find valuable information regarding past and current research. Many questions you have are addressed in the FAQs tab. In the Blog, you will read first hand accounts from KAT6A families who may be going through a similar experience as you. You will also find news articles and videos featuring our children in the Blog. You can find out about upcoming fundraising and social events in our KAT6A community by visiting the Events page. Furthermore, you can help us raise money for research by encouraging your friends and family to visit our Donation page. The website is updated regularly, so please come back often and share it with your medical specialists and family members.
Our 501(c)(3) nonprofit organization was started in 2017 by several dedicated parents from our support group. We strive to help families coping with a KAT6A diagnosis by providing resources and support. An important part of our mission is to raise funds for research so that we can learn more about the KAT6A gene, find treatments or a cure. The KAT6A Foundation also aims to raise awareness of this rare disorder so that more individuals are diagnosed. We also plan to start a registry. As more money is raised, we will directly help families in need of financial help to acquire adaptive medical devices and other treatments not covered by insurance.

If you are interested in fundraising, please share our web site, www.kat6a.org, through email or on Facebook. Our foundation is run entirely by volunteers, so 100% of donations will go towards research. We also welcome you to fundraise on Facebook by choosing KAT6A Foundation, Inc for your birthday charity. Furthermore, you can help us raise funds by shopping on www.smile.amazon.com. Read our quarterly newsletter to get updates on fundraising totals and where donations are going. Help us reach our goal of $100,000.

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.

Please like/follow/subscribe to us on social media. Invite your friends, too!

Facebook
https://www.facebook.com/kat6afoundation/

Instagram
https://www.instagram.com/officialkat6a/

Youtube
https://www.youtube.com/channel/UCPJcgZbhzqGZq0k2VRIn9LQ

We are eager for your help and to hear your ideas. If you’d like to get involved with the foundation and help on one of our committees, we welcome you to join our KAT6A Foundation Fundraising and Awareness Group today. https://www.facebook.com/groups/1924074790952872/
**FAQs**

**My child was just diagnosed with KAT6A, what medical steps should I take?** It is recommended that every individual should see a cardiologist to rule out any structural heart problems. It is also important to have vision assessed by a specialist. If your child does not already receive speech therapy, it is important to start now.

**What treatments are available?** Currently doctors are treating the symptoms related to KAT6A on an individual basis, but there is no medication for KAT6A specifically. It is important to start therapies that target different developmental delays through early intervention programs and special education services as early as possible.

**What causes KAT6A?** KAT6A syndrome arises from a spelling mistake in the KAT6A gene. The majority of KAT6A mutations are de novo, meaning that it was a new change in the gene, not inherited from either parent and there is nothing either parent did to cause this.

**How many people have KAT6A?** Through January 2018, there are 113 known cases, but we believe many more individuals will be identified as exome sequencing becomes more common.

**Where are KAT6A individuals living around the world?** KAT6A individuals have been identified in many countries including: Argentina, Australia, Austria, Belgium, Canada, Chile, China, Dominican Republic, England, Finland, France, Germany, Ireland, Israel, Japan, Netherlands, Norway, Scotland, Spain, Sweden, Venezuela, and the United States.

**How can I inform others about KAT6A?** Your best reference tool is our KAT6A Foundation website, [www.KAT6A.org](http://www.KAT6A.org). The web page is a compilation of all the research. It includes the most current information on KAT6A, and is updated regularly. In addition to our web page, Unique created this helpful handbook for families. We recommend printing a copy for each of your child’s medical specialists and therapists. [https://www.rarechromo.org/media/singlegeneinfo/Single Gene Disorder Guides/KAT6A Syndrome FTNW.pdf](https://www.rarechromo.org/media/singlegeneinfo/Single Gene Disorder Guides/KAT6A Syndrome FTNW.pdf)

**Will my child learn to talk?** All KAT6A individuals have language delays. Yet, there’s a wide range in language ability in this group. Some children are nonverbal and communicate through signing, body language, PECS (picture exchange system) or adaptive technology. Others are verbal teens and adults despite language delays as young children. Many parents report that their children have markedly better receptive language than expressive language.

**What is the likelihood I could have another child with KAT6A syndrome?** In de novo KAT6A gene mutations, there is a 1-3% risk of KAT6A syndrome reoccurring for the same parents.
What therapies should my child be getting? Every individual is different, but many in the KAT6A community receive a wide variety of therapies as children to aid in their development.

- Physical therapists help improve gross motor development, which is typically delayed in our children due to abnormal muscle tone and poor coordination.
- Occupational therapists work on fine motor activities required for daily living.
- Speech therapists work with our children in developing speech production, receptive language, signs and vocalizations. Many KAT6A children have apraxia and struggle with the motor planning required for fluent speech.
- ABA therapy can be a successful method for children who have autism.
- Feeding therapists work with children with feeding difficulties. Many KAT6A children have feeding delays due to hypotonia and acid reflux, and others have structural damage that require feeding tubes.
- Vision therapy is beneficial for individuals with CVI (Cortical Visual Impairment) or strabismus.
- Special education accommodations will likely be required for your child. Individuals with KAT6A have a wide range in intellectual ability, so it is impossible to predict how your child’s needs will be met in school. Early intervention programs may offer a special educator starting at birth if cognitive delays are present.
- Various alternative therapies may benefit your child, such as: aquatic therapy, music therapy, sensory therapy, equine-assisted therapy, CME therapy, and biofeedback.

What is all this talk of a mitochondrial cocktail in the support group? Some parents have observed benefits from a mitochondrial cocktail that contains different over the counter supplements and vitamins. The main components of the cocktail are carnitine and vitamin B5. It is essential to consult your child’s physician before starting any new supplements. Dr. Richard Kelley did a web presentation for our group on the possible benefits for KAT6A individuals. You can find the slides of his presentation in the “Files” section of the support group page at Facebook https://www.facebook.com/groups/803280496369674/.

If you have further questions, you can send a private message to Natacha Esber, a parent in our support group who is also a medical doctor and has a wealth of knowledge on this topic. It is important to keep in mind that a cocktail is not a cure and may have no significant improvements for your child. Each family has made their own personal decision whether it is the right choice for them.
KAT6A Family Resources

May 2018

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Contact us at support@kat6a.org with any questions or comments. We are here to serve you.
Inspirational Achievements

There is a wide range in abilities in the KAT6A population. We are proud of all of our kids and adults and wanted to share some of the highlights with you.

Four-year-old Ginger is having success on skis.

13-year-old Cullen made his town’s swim team. He has been competing in Special Olympics for many years, but now he will be competing against typically developing children.

One KAT6A individual received his driving permit at age 16.

There are only a handful of adults diagnosed with KAT6A, but two known individuals have part-time jobs.

Several individuals communicate very well using a speech communication device or by signing.

Eight-year-old Peter’s literature entry recently won the Reflections PTA Artist for his school, then his county, and the statewide competition for New York. Despite being nonverbal, Peter understands and can translate by typing 4 languages. He also plays piano and can sight read music sheets.

So many of our kids just make everyone around them smile. Most KAT6A individuals have a loving and happy disposition and can light up any room.

Several children have learned to ride a bike, ski, swim and play various other sports.

Beckem, age 5, loves to learn. He can identify various organs of the body.

John, age 12, enjoys fishing, playing basketball, and participates in many events in the Special Olympics. He loves math, geography and writing essays in his mainstream sixth grade classroom.

Nine-year-old Chloe is a chatterbox and will carry on a great conversation. There are several other verbal KAT6A children. This fact you will not find in any of the research articles.

Sam, age 27, has never spoken and doesn’t appear to have any self-help skills, but he gives the very best hugs, the best hand massages, shows infectious enthusiasm with huge smiles, and has a talent for making people forget their worries and cares. That is a gift.
# KAT6A Providers List

**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
I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met. It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever go away... because the loss of that dream is a very very significant loss.

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.