# About Us

The KAT6A Foundation was founded in 2017 by parents of children identified with mutations on their KAT6A gene. At the time, there were fewer than 50 known KAT6A cases. In 2020, we began fostering connections with the KAT6B community and in 2022, we formally became the KAT6 Foundation, an organization devoted to understanding mutations in both KAT6A and KAT6B genes. Today, there are more than 500 known cases of KAT6 syndromes worldwide, and we expect our numbers to continue to grow.

We are the only 501(c)(3) nonprofit organization founded to support the international KAT6 community.

# Our Mission

The KAT6 Foundation supports individuals and their families who are living with KAT6A and KAT6B syndromes around the world. We advance scientific research aimed at developing treatments, and spread awareness of KAT6 syndromes so they can be more easily identified, treated, and studied.

"Alone we can do so little; together we can do so much."

KAT6FOUNDATION.ORG


# Join us

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KAT6 Foundation

# Driving Research

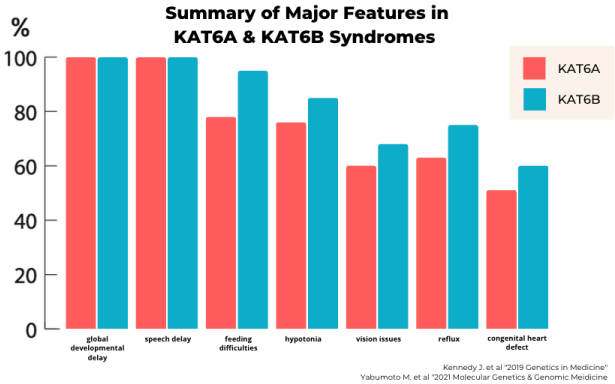
- Chan Zuckerberg Initiative, USA - the KAT6 Foundation was chosen as part of the CZI: Rare as One Network — a group of 30 patient-led organizations that are accelerating research and driving progress in the fight against rare disease
- Center for Regenerative Medicine (CREM), USA - KAT6 Foundation and collaborators have established the first patient-derived induced Pluripotent Stem Cells (iPSC) bank for KAT6A and KAT6B variants
- Centro Andaluz de Biología del Desarrollo (CABD), Spain – first research study directly funded by our foundation was published by Genes on Nov 15, 2022, *Pantothenate and L-carnitine Supplementation Corrects Pathological Alterations in Cellular Models of KAT6A Syndrome*
- David Geffen School of Medicine, USA - stem cell research on KAT6 variants
- Technion Institute, Israel - multiplex analyses of the molecular pathophysiological mechanisms underlying KAT6
- Walter and Eliza Hall Institute of Medical Research, Australia - KAT6 mice studies are underway with potential treatments being tested
- Murdoch Children's Research Institute, Australia - speech study on KAT6
- Kennedy Krieger Institute, USA – study of cognition and neurobehavior in KAT6
- Rare Diseases: Models & Mechanisms Network (RDMM), Canada- study involving fly models to investigate the neurological implications of KAT6

# Supporting Families

- The KAT6 Foundation's Empowered Grant program provides funding for the purchase of assistive equipment, devices, and technology, as well as a variety of therapies for individuals diagnosed with KAT6 syndromes.
- Our advocacy team helps caregivers navigate the educational and medical systems to make informed decisions and obtain optimal services and care for their child.
- We are the official sponsor of the annual KAT6 Conference. Each conference enables open dialogue between families, clinicians, and researchers and provides a platform for the KAT6 community to expand its network.

# Features of KAT6

Each person with KAT6 syndrome has a different mutation along the KAT6A or KAT6B gene, which leads to a wide range in symptoms and features.




Our KAT6A/KAT6B Patient Registry launched in 2019 through the National Organization of Rare Diseases (NORD). Our registry collects valuable data about many aspects of KAT6 syndromes, enabling researchers to understand the full range of KAT6 characteristics and to identify areas for additional studies.