KAT6 Foundation

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To support individuals and their families who are living with KAT6A and KAT6B syndromes around the world.

KAT6 syndromes are ultra-rare diseases caused by mutations in the KAT6A and KAT6B gene, with common symptoms including global developmental and speech delays, feeding difficulties and hypotonia. For the approximately 500 people diagnosed with KAT6 gene variants, treatment is directed toward the specific symptoms that are apparent in each individual, but no specific medications for KAT6 syndromes exist.

Peter Najm, who has the rare disease KAT6A Syndrome, presented his poster about genes and DNA to school management alongside his siblings and parents.

During the Grant Period

Impact Spotlight

When the organization led by Emile Najm received funding to join the Rare As One Network in 2019, it was known as the KAT6A Foundation. Since its founding in 2017, the organization and its small, ultra-rare KAT6A community had struggled to attract industry interest or investment.

At an early patient-research convening organized by the foundation — for some scientists, their first opportunity to meet a patient with KAT6A syndrome — a researcher encouraged the organization to expand its remit to include KAT6B syndrome, a disease with many significant overlaps to KAT6A syndrome. Though an exciting chance to approximately double the patient community and expand the organization's impact, a broadening of scope was not without challenges, including financial costs and mission expansion requiring greater investment and expertise.

Nevertheless, over the next several years, the KAT6A Foundation would commit to making these investments, gradually building relationships and trust. By the end of the grant period in 2023, the officially-renamed KAT6 Foundation represented a combined registered patient community of 187, with KAT6B syndrome representation on its board, and a goal of ultimately achieving 50% representation from KAT6A and KAT6B leaders.

For the KAT6 Foundation, this value of inclusion has remained central throughout their work. Additionally, the organization has broadened its international scope by involving researchers in Europe, and also expanding its reach to include patients in Australia.

Key research and research infrastructure achievements

- Provided seed funding to support creation of an iPSC line that covers many variations of the KAT6A and KAT6B gene mutations.
- Created a sharing policy among members of the collaborative research network, to promote open science.
- Supported research that confirmed the effectiveness of a vitamin therapy, utilizing fibroblasts obtained from patients.

Key publications

- *Human Genetics and Genomics Advances* (2021): a cross-disorder study describing 19 novel episignature disorders and comparing findings alongside 38 previously established episignatures for a total of 57 episignatures associated with 65 genetic syndromes, providing insight into the molecular etiology of Mendelian conditions like KAT6 (co-authored by KAT6 Foundation co-founder Natacha Esber).
- *Genes* (2022): a study on the pathogenic variants of the KAT6A gene, and the therapeutic effectiveness of epigenetic modulators and mitochondrial boosting agents (funded by the KAT6 Foundation).

Key operational achievements

- Hired a Fundraising Manager, who helped increase funds raised from $75,000 in 2019 to close to $375,000 in 2022.
- Hired a research coordinator in Australia.

Key community achievements

Expanded patient and research communities, growing conference attendance from 19 families and three speakers in 2018 to 180 attendees and 17 researchers/speakers in 2023.

After the Grant

- In 2023, the KAT6 Foundation established a committee to study patient mortality, with a goal to guide parents in understanding how best to adjust to KAT6 disorders and prevent suffering among the most vulnerable members of the patient community.
- Additionally, the foundation expanded the availability of its KAT6 Handbook, a resource for parents, teachers, doctors and caregivers, to seven languages, including Arabic, French, Italian and Portuguese.
- The foundation continues its work to engage both KAT6A and KAT6B patients. In 2024, the KAT6A Patient Registry was expanded to include all KAT6 families, and renamed accordingly, to the KAT6A/KAT6B Patient Registry.
- In June 2024, the foundation convened its community for the 5th International KAT6A & KAT6B Conference, attended by more than 70 families and 18 researchers.

Angie Serrano (left) and Saylor Williams (right), of Boston University, attend the Science in Society 2023 Meeting as research partners of the KAT6 Foundation. Dr. Serrano leads a research program that uses iPSC-derived models and zebrafish to understand cardiovascular and neurodevelopmental manifestations in rare disorders.

"Families are close to my heart. Witnessing someone in pain reminds me of my son's struggles. I am determined to spare others from experiencing such hardships."

Emile Najm
Co-Founder and CEO, KAT6 Foundation