The KCNT1 Epilepsy Foundation's mission is to support the community and to accelerate research and drug discovery efforts through multiple initiatives:

- Patient registry
- Invitae digital natural history study
- Biobank - including patient iPSCs
- Community education
- Fundraising and research grants

**The Science**

- Gain of function K+ ion channelopathy
- De novo & autosomal dominant missense variants

**Common Clinical Manifestations**

- Drug resistant seizures
- GI / GU issues
- Ketogenic diet success
- Motor involvement
- Sleep disturbances
- Autism features

**Phenotypes**

**EIEE/EOEE:** Early-onset infantile epileptic encephalopathy
- Onset < 6 months
- Infantile Spasms
- Global Delay

**EIMFS/MMPSI:** Migrating focal seizures
- Onset < 6 mos
- Limited neurodevelopment
- Respiratory compromise
- Feeding & swallowing issues
- CVI and vision issues
- Hypo and hypertonicity
- Neurogenic crying
- Regression or stagnation
- Cardiac disorders (MAPCAs)
- SUDEP risk

**ADNFL:** Autosomal dominant nocturnal frontal lobe seizures
- Onset > 6 months
- Sleep hypermotor activity
- Psychiatric disorders
- Learning disabilities
- Misdiagnosed as night terrors

**Therapeutic Strategies**

- ASOs, RNAi, gene editing
- Small molecule development
- Drug repurposing

**Connect with KCNT1**

EIN 84-2748218
https://kcnt1epilepsy.org
We provide an important role in bringing families, clinicians and researchers together to ensure the patient perspective is included in the research and drug development process. We encourage the scientific community to collaborate to understand KCNT1 gene variants, and help fund research necessary to develop multiple treatment options -- and find an eventual cure for KCNT1-related disorders.

Supporting Families
Caring for a loved one with any health issue can impact multiple areas of a family's life. It is important to find a support system that consists of family, friends, other parents with KCNT1 experience, therapists, and also respite care. Contact us and we can connect you with other parents, and invite you to our private support groups. You can also benefit from our educational webinars and find resources on our website.

Treatment
There are no specific treatments for KCNT1-related disorders today. Instead, the goal is to minimize seizures, treat symptoms, and provide opportunities to support development and prevent loss of skills. There are several drugs currently in development and we are hopeful these will be available in the near future. Children with early onset seizures benefit from a multidisciplinary care team including physical, speech and vision therapies. Children with later-onset KCNT1 and nocturnal seizures can have autism spectrum disorder or autistic-like features and behavioral issues. Early intervention is important. More information is available on our website at https://kcnt1epilepsy.org

Thank You for Your Support
We are a nonprofit organization recognized by the United States Internal Revenue Service (IRS) also known as the KCNT1 Slack Epilepsy Foundation. In order to get closer to a cure for KCNT1-related disorders, we need your support. You can feel confident that your gift is carefully stewarded by the KCNT1 Epilepsy Foundation Board of Directors. We are run by volunteer family members and aim to be transparent and operate with minimal expenses. Please know your contributions support our mission of improving the lives of those living with KCNT1 life-threatening disorders by funding research necessary for treatment development.

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