WHO WE ARE

- We are a global parent-led nonprofit dedicated to raising awareness, supporting families, and finding treatments.
- Our mission: To educate, advocate, and advance research toward effective treatments and a cure.

HOW YOU CAN PARTICIPATE

Support Us – Donate, host/attend fundraisers, join our network.

Get Involved – Volunteer, share your story, join our email list.

Advance Research – Participate in studies, attend our conference

KCNQ2 AWARENESS WEEK & INTERNATIONAL AWARENESS DAY

KCNQ2 Awareness Week is held annually from March 1-7, and KCNQ2 International Awareness Day is March 2nd.

What does KCNQ2 stand for?

- **K** The chemical symbol for "Potassium"
- **CN** Stands for the word "Channel"
- **Q** Sub family of the potassium channel
- **2** 2nd member of the "Q sub-family of K channels"

FOLLOW US AND JOIN OUR COMMUNITY

KCNQ2cure.org

info@kcnq2cure.org

Links available on website:

KCNQ2 Cure Alliance

KCNQ2 Cure Alliance

Unity. Hope. Strength. Love

Understanding KCNQ2. **Supporting Families. Inspiring Hope.**

LOVE

WHAT ARE KCNQ2-RELATED DISORDERS?

KCNQ2-related disorders are a rare form of developmental disorders that can begin as early as birth and can significantly impact people throughout their lifetime. KCNQ2 itself is not a medical condition; rather, it is the name of the gene that undergoes a pathogenic change.

Variations in the KCNQ2 gene are responsible for a range of neonatal-onset epilepsy disorders, encompassing both severe early-onset epilepsies known as developmental and epileptic encephalopathies (DEEs), as well as milder forms referred to as self-limited neonatal epilepsies, which may run in families. The impact of these genetic variations can vary widely, resulting in diverse symptoms and varying degrees of severity, depending on the specific KCNQ2-related disorder a child has.

OTHER POSSIBLE CHALLENGES

Every child's journey is unique. Not all children with KCNQ2-related disorders will experience these conditions, but some may include:

- Learning or speech delays
- Movement or motor difficulties
- Autism or behavioral concerns
- Sleep or GI problems
- Vision or orthopedic challenges

RESOURCES

KCNQ2 GLOBAL SUPPORT NETWORK

The KCA Global Support Network is a program designed for parents, offering online support groups, educational seminars, and family meetups.
Scan the QR code to connect.

CAREGIVER CONNECT

A monthly virtual meet-up with no agenda, just time to talk, listen, and share with others who truly understand the KCNQ2 journey. *Scan the QR code to join us.*

CAMBRIDGE ELEMENTS - GENETICS IN EPILEPSY

The Cambridge Elements guide, created with top researchers, helps families understand KCNQ2-related epilepsy and its wide range of symptoms. It explains how genetic changes affect development and treatment, giving families clearer answers after diagnosis.
Scan the QR code to access.

KCNQ2-RELATED DISORDERS

KCNQ2-DEE. Seizures may be resistant to multiple anti-seizure medications. Seizure freedom is more likely to be achieved when receiving medications known as sodium channel blockers. Seizures generally disappear in childhood, and individualized therapies are recommended for developmental delay and intellectual disability.

Variants in the KCNQ2 gene can disrupt the potassium channel in two main ways: **Loss of Function** (LoF) or **Gain of Function** (GoF).

- A LoF variant reduces the excitability or destroys channel function altogether, typically leading to neonatal seizures and/or developmental and intellectual disability..
- A GoF variant makes the channel more excitable, typically leading to developmental and intellectual disability.

Less common types of KCNQ2-related disorders:

- Neonatal encephalopathy with non-epileptic myoclonus
- Non-neonatal-onset developmental and epileptic encephalopathy
- Intellectual disability (without seizures).

UNITY, HOPE & STRENGTH