NAVIGATING KCNQ2

A Guide for Families and Caregivers

KCNQ2 Cure Alliance

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About KCNQ2 Cure Alliance (KCA)

KCNQ2 Cure Alliance was founded in 2015 by a group of parents determined to make a difference. Like many families, we faced a diagnosis we had never heard of and a future full of questions. We came together with a shared purpose: to raise awareness, support families, and accelerate the path to better treatments and a cure.

Since then, we've grown into a dedicated community of caregivers, researchers, and advocates. Our team remains parent-led and volunteer-driven. Every effort we make is rooted in the lived experiences of families navigating KCNQ2-related disorders (KRDs).

Thanks to the support of our community, we have funded cutting-edge research, fostered global collaboration, and created educational tools for families, caregivers, and clinicians. From launching research studies to hosting family conferences, we remain committed to progress and connection.

We invite you to stay connected, get involved, and help advance this mission. Together, we are **supporting families, advancing research, and channeling hope.**

Welcome to the KCNQ2 Community

Receiving a diagnosis of a KCNQ2-related disorder (KRD) can feel like the ground has shifted beneath you. Grief, fear, confusion, and feeling overwhelmed are all valid and common responses. Many of us in this community have stood exactly where you are now. That doesn't make it easier, but it means you don't have to figure it out alone.

KCNQ2 Cure Alliance was created by families, for families. We have walked this path too, and we are here to offer information, support, and connection as you begin navigating life with KCNQ2.

This guide was created with input from caregivers, clinicians, and advocates. It doesn't have every answer, but it offers a starting place. You'll find:

- Overview of KCNQ2-related disorders
- What to expect after diagnosis
- How to manage seizures, therapies, and daily life
- Information about the KCNQ2 gene and genetics
- A look at current research and how to get involved

Every KCNQ2 diagnosis is different. Every family's journey is unique. There's no one-size-fits-all plan, and that's okay. Take what's helpful now and return to the rest when you're ready.

Most importantly, remember this: **you are part of a strong, compassionate community that understands**. We are here with you, every step of the way.

How to Use This Guide

You don't have to read everything at once.

Start with what feels most relevant, and come back as new questions arise. Each section is designed to stand alone, so you can move between practical information, emotional support, and scientific context as needed.

If you're reading digitally, use the Table of Contents to jump directly to the section you need.

Quick Start Guide

Getting a diagnosis can feel overwhelming, but you don't have to figure it all out at once. This quick-start checklist is here to help you take the first steps at your own pace. If there's anything on this list you're not familiar with, don't worry. You'll find more information in the pages of this guide.

Medical & Diagnosis

- Schedule an initial or follow-up visit with a neurologist
- Request a copy of your loved one's **genetic report**.
- Ask your neurologist to refer you to a genetic counselor and a developmental pediatrician.
- Work with your neurologist to create a **Seizure Action Plan**.
- Fill and carry your loved one's **rescue medication**, and learn exactly when and how to use it.
- Start logging your loved one's seizures, symptoms, and milestones using a journal or an app.
  - **Resource**: Seizure Tracker

Emotional & Community Support

- Give yourself permission to feel everything — grief, fear, hope, love.
- Set aside time to talk with a friend, counselor, or another KCNQ2 parent about how you're feeling.
- Ask loved ones how they can help (meals, childcare, errands, rides).
- Schedule one **restful moment for yourself this week**, even if it's just 15 minutes.
- Bring a friend or partner to appointments for support and note-taking.
- Join the private Facebook group, **KCNQ2 Global Support Network for Caregivers**, to connect with others.
- Subscribe to the **KCNQ2 Cure Connections Newsletter** at kcnq2cure.org.
- Write down one small act of self-care you'll try this week — and do it.

Quick Start Guide Continued

Therapies & Development

- Ask your pediatrician or care team for referrals to **physical, occupational, speech, and vision therapy** as needed.
- Discuss feeding, gastrointestinal, or sleep issues with your doctor and request specialist referrals when necessary.
- Explore supportive therapies such as **aquatic therapy, music therapy, or equine therapy** to help with development and comfort.

You may also want to explore specialized and intensive therapy options that can help your loved one make meaningful progress in shorter periods of time:

- Therapy Intensives
- DMI (Dynamic Movement Intervention)
- Conductive Education

Care Planning & Organization

- Begin a **medical notebook or digital folder** to track medications, appointments, symptoms, and questions.
  🔗 **Resource**: How to Build a Medical Binder
- Track dates for starting/stopping medications, new concerns, and milestones (or “inchstones”).
- Keep an updated list of medications and emergency contacts handy for caregivers and schools.

Tip
You don’t need to be perfect to be a great advocate. Every note you take, every question you ask, and every act of love you show matters more than you realize.

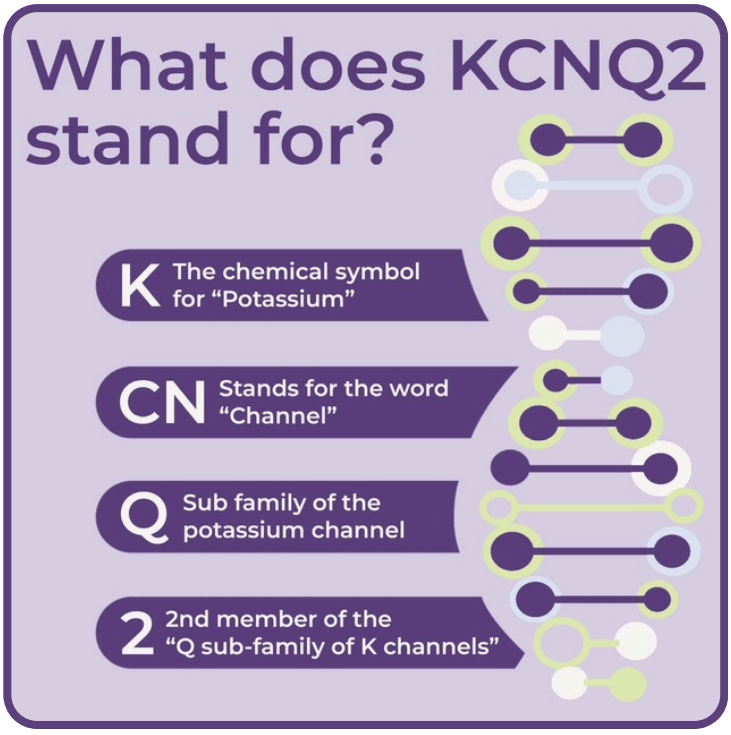
What KCNQ2 Means for Your Family

As genetics advances rapidly, even experts can struggle to keep up with the "alphabet soup" of gene names. Different testing companies and medical teams might use slightly different terms, but they all fall under the umbrella of KCNQ2-related disorders (KRDs).

What's in a Name?

With genetics advancing rapidly, it is challenging for everyone to keep up with the names of all the genes in the alphabet soup. Even genetic testing companies use different terms, but they all fall under the umbrella of **KCNQ2-related disorders (KRD)**. You might see one of these terms on a report or in a clinic note:

- Developmental and Epileptic Encephalopathy-7
- KCNQ2 Encephalopathy
- KCNQ2 Epilepsy
- KCNQ2 Developmental and Epileptic Encephalopathy (DEE)
- KCNQ2 Neurodevelopmental Disorder
- KCNQ2 Self-Limited Familial Neonatal Epilepsy (SLFNE) — formerly called Benign Familial Neonatal Epilepsy (BFNE)



The KCNQ2 Spectrum

KCNQ2-related disorders exist on a **spectrum**. Some individuals experience seizures that resolve in infancy and have little or no developmental impact. Others face ongoing challenges with movement, communication, learning, or health.

Understanding where your loved one fits on this spectrum can help set expectations and guide care, but it's important to remember that no single description captures every child.

Two Common Diagnostic Terms

- **Developmental and Epileptic Encephalopathy (DEE)** or **KCNQ2-Related Epilepsy**
  - Often involves ongoing developmental delays or disabilities.
  - Seizures may be difficult to control or may return later in life, even after a seizure-free period.

- **Self-Limited Neonatal Epilepsy (SeLNE)**
  - Seizures begin in the first days of life, but typically resolve in infancy.
  - Development is often typical or only mildly affected.
  - Often, there is a positive family history of seizures in infancy in a parent or other family member.

Both fall within the **KCNQ2 spectrum** and share the same underlying cause — a change in the KCNQ2 gene.

Tip
Remember: a diagnosis is a guide, not a prediction. It helps doctors coordinate care and services but doesn't define your child's potential.

One Mutation, Many Outcomes

A KCNQ2 diagnosis is an important starting point, but it doesn't tell the whole story. Even individuals with the same change in the KCNQ2 gene — known as a **variant** or **mutation** — can develop in very different ways.

Some children may walk and talk independently, while others use wheelchairs or communication devices. Some have long stretches without seizures, while others face ongoing health challenges.

Doctors look at both your loved one's genetic test results and their unique symptoms to guide care. Even then, it's impossible to predict the exact path ahead.
There is no crystal ball. And while that uncertainty is difficult, it also leaves room for **growth, joy, and hope**.

Even within a KCNQ2 diagnosis, there's a wide spectrum of symptoms and outcomes. Every individual is unique — even those who share the same gene variant. It's best to approach the diagnosis with **balanced hope**: stay informed about what might arise, but remember that your loved one's path will unfold in their own way.

It's natural to want concrete answers. But KCNQ2-related disorders are still being studied, and our understanding continues to evolve. Over time, researchers may redefine subtypes and clarify how different genetic changes affect outcomes.

Think of a diagnosis as a **tool for coordination**, not a rigid definition. It can help you access therapies, services, and medical support, even if your loved one's symptoms don't fit neatly into a single category like epilepsy, autism, or cerebral palsy.

Your participation in research and community efforts helps fill in these missing pieces. **Every family's story contributes to progress and a clearer future for KCNQ2**.

Tip
Consider joining a registry or research platform such as KCNQ2 Cure Research at Citizen, to share your family's data and experience.

Changes Over Time

Many families find that symptoms evolve as their loved one grows. Some children outgrow seizures completely, while others may continue to experience them.

Movement, communication, and feeding abilities may improve, plateau, or become more complex with age. These shifts can feel unpredictable — but they also show that development is dynamic and ongoing.

Your loved one may meet **milestones** or **inchstones** on their own timeline and in their own way.

- *Milestones* are the big moments — walking, talking, eating independently.
- *Inchstones* are the smaller steps that matter just as much — holding eye contact longer, smiling at a familiar voice, reaching for a toy.

These quiet victories often bring families deep joy, even if they aren't always measured by standard tools.

As a parent or caregiver, it's normal to feel overwhelmed. You don't need to have all the answers. By showing up with love, care, and curiosity, you're already doing something powerful.

Take it one day at a time. Ask questions. Celebrate small wins. And remember: there's a whole community of families, doctors, and researchers walking this path beside you.

If you're curious about **what causes KCNQ2** or want to understand your loved one's genetic results, you'll find clear explanations later in this guide under Section 4: Understanding the Science Behind KCNQ2.

Understanding KCNQ2: The Basics

KCNQ2-related disorders (KRDs) are complex neurological conditions that affect how brain cells communicate.

While seizures are often the first sign, KRDs can influence many areas of development and daily life.

Common challenges may include:

- Motor delays or low muscle tone
- Speech or communication difficulties
- Learning and cognitive differences
- Sleep disturbances
- Gastrointestinal issues (such as reflux or constipation)

Researchers estimate KCNQ2 mutations may occur in roughly **1 in 17,000 births**, meaning thousands of children worldwide are likely affected, but not yet diagnosed.
KCNQ2 mutations alter how potassium channels in brain cells open and close, changing the way neurons send signals.

These changes can lead to seizures and differences in development, movement, or behavior.

Seizures and KCNQ2

Early Seizures

For most families, seizures are the first visible sign that something is different.

Seizures in KCNQ2 often begin in the first days or weeks of life.

- Many children's seizures can be controlled with medication and may stop in early childhood.
- Some continue to have seizures or see them recur later in life.
- Others never experience seizures at all.

Every child is unique — and so is their response to treatment.

What to Expect Over Time

Seizure patterns and symptoms may change as your child grows.
Some gain skills steadily; others face longer plateaus or more complex needs.
Remember: progress looks different for every family.

Managing Seizures

Effective seizure management requires teamwork between you, your neurologist, and other care providers.

Common Seizure Types in KCNQ2

Most individuals with KRDs begin having seizures in the first days or weeks of life. It is important to understand the type of seizure(s) your loved one is experiencing. These may include:

- **Tonic seizures** – body stiffening
- **Tonic-clonic seizures** – stiffening followed by jerking
- **Focal seizures** – affecting one part of the body
- **Myoclonic seizures** – quick, sudden muscle jerks
- **Atypical absence seizures** – brief staring spells or blinking
- **Status epilepticus** – seizures lasting longer than five minutes or happening back-to-back without recovery; these require emergency medicine
- **Infantile Spasms**: Brief, repetitive contractions (sometimes seen with hypsarrhythmia on an EEG)

Seizure Resources

Creating a Seizure Action Plan (SAP)

A written SAP helps everyone who cares for your loved one — family, teachers, and medical teams — respond safely during a seizure.

Include:

- Seizure types and patterns
- When to give rescue medication
- Emergency steps and key contacts

📄 **Template**: Seizure Action Plan Form

Rescue Medications

If a seizure lasts too long or does not stop on its own, rescue medication (e.g., Diazepam, Midazolam) may be needed.

- Your neurologist will prescribe what's right for your child.
- Keep it with you and ensure others know how and when to use it.
- Replace expired medication promptly.

Safety and SUDEP Awareness

SUDEP (Sudden Unexpected Death in Epilepsy) is rare, but important to understand.

To reduce risk:

- Take medications as prescribed.
- Act quickly if a seizure lasts longer than five minutes.
- Monitor during sleep when possible.
- Discuss SUDEP openly with your care team.

🔗 **Resource**: Epilepsy.com SUDEP Toolkit

Tools and Resources

Monitoring Tools at Home

Many families find peace of mind using simple tools to track sleep or movement:

- Video monitors: Standard infant cameras
- Movement monitors: Detect shaking or twitching
- Pulse oximeters: Track oxygen and heart rate
- Wearable devices: Smartwatches or patches that detect seizure patterns

Common Treatments and Medications

There is no single drug that works for everyone with a KRD, but many achieve control with anti-epileptic drugs **(AEDs)**.

- **Sodium channel blockers** often help with loss-of-function mutations*.
- **Other approaches** may be needed for gain-of-function variants.
- Treatment plans should consider both genetics and symptoms.

*(Loss-of-function and gain-of-function are described later in Section 4: Understanding the Science Behind KCNQ2)

Additional options include:

Ketogenic Diet – A high-fat, low-carb diet that can reduce seizures for some. A neurologist and a dietitian must supervise it.

Vagus Nerve Stimulation (VNS) – A small device implanted under the skin sends gentle electrical signals to the brain through the vagus nerve. For those who don't respond well to medication, VNS may reduce the frequency or severity of seizures.

Tip

No device is foolproof. Use technology as an aid — not a substitute for careful observation and medical guidance.

Beyond Seizures: Other Health Challenges in KCNQ2

KCNQ2-related disorders often begin with seizures but can influence many other aspects of health and development.

Some children experience multiple challenges; others only a few. Early, consistent intervention can make a meaningful difference.

🧠 Development, Cognition, & Communication

- Developmental delays in speech, movement, and learning are common.
- **Speech therapists** support communication, feeding, and safe swallowing.
  - Tools may include picture cards or Speech-Generating Devices (SGDs).
- **Occupational therapists** assist with skills such as grasping, feeding, dressing, and sensory integration.
- **Physical therapists** focus on strength, balance, coordination, and mobility.

Early intervention through school or community programs is key — start as soon as possible and re-evaluate often.

💪 Motor and Movement Differences

Some individuals experience:

- Low muscle tone (hypotonia)
- Stiff muscles (spasticity) or involuntary movements (dystonia)
- Coordination difficulties (ataxia)

Support strategies include:

- Ongoing physical therapy for core strength and posture.
- Adaptive tools such as orthotics, walkers, or adaptive strollers.
- Play-based activities like aquatic therapy or hippotherapy (horseback riding).

Feeding and Gastrointestinal (GI) Challenges

- Common concerns include constipation, reflux, and feeding difficulties.
- Feeding therapists (usually speech or OT specialists) help build safe mealtime skills.
- Gastroenterologists address reflux, motility, and nutrition issues.
- Some children benefit from feeding tubes (G-tubes) to support growth, hydration, and medication delivery.

Sleep Differences

Difficulty falling or staying asleep is common in KCNQ2.
Possible contributors include seizures, reflux, or neurological differences.

- Discuss safe options with your doctor (e.g., **melatonin** or other sleep aids).
- Maintain consistent bedtime routines and sleep environments.
- Many families use infant monitors, pulse oximeters, or wearables for nighttime monitoring.

Behavior & Sensory Processing

Some individuals experience strong emotions, difficulty with attention, or sensory sensitivities to sound, light, or touch.

- Occupational therapy helps children tolerate and organize sensory input.
- Behavioral therapy (ABA or play-based) can support communication, routines, and emotional regulation.
- Developmental pediatricians or child psychologists can help design behavior and medication plans.

Other Medical Needs

Some children also experience:

- **Cerebral Visual Impairment (CVI)** or other vision challenges
- **Neuropathic pain** or muscle tightness
- **Dysautonomia,** where the autonomic nervous system does not function properly

You may work with:

- Functional vision specialists
- Audiologists and ophthalmologists
- Neurologists and geneticists for long-term planning

Caring for the Caregiver

When your loved one is diagnosed with a KRD, life can quickly fill with appointments, paperwork, and new responsibilities.

Your well-being is not secondary — it's central to your child's care.

Simple Ways to Care for Yourself

- **Ask for help.** Let friends and family pitch in with meals or errands.
- **Give yourself grace.** No one has all the answers. Do your best and rest when you can.
- **Find your people.** Connect with others in the KCNQ2 community or local support groups.
- **Take micro-breaks.** Five minutes of fresh air or quiet can re-set your energy.
- **Prioritize sleep and nutrition.** They are the foundation of resilience.
- **Move your body.** Stretch, walk, dance — whatever feels good.
- **Keep a journal.** It can track medications, appointments, and your own thoughts.
- **Nurture relationships.** Make time for your partner or close friends.
- **Seek help early.** Therapy or peer support can prevent burnout and build coping skills.

Caring for the Caregiver: How to Recognize and Address Parent and Caregiver Burnout

You don't have to be strong all the time to be an amazing caregiver.
Caring for yourself is part of caring for your child.

Talking to Siblings, Family, & Friends

A KCNQ2 diagnosis affects the whole family. Siblings and extended relatives may have questions, confusion, or big feelings they don't know how to express.

Open conversations build understanding and connection.

Tips for Healthy Communication:

- Use simple language: "Your brother's brain works a little differently, so he needs extra help from doctors and therapists."
- Acknowledge emotions: Let siblings know it's okay to feel sad, angry, or confused.
- Share updates: Include family and friends so they feel connected and confident helping.
- Make special time: Plan "just us" moments with siblings to show they matter too.
- Encourage questions: No question is too small or off-limits.

If siblings struggle with big feelings, counseling or peer groups can help them process their experience.

What Is a Genetic Mutation?

Think of your DNA as a giant **cookbook** filled with thousands of recipes.

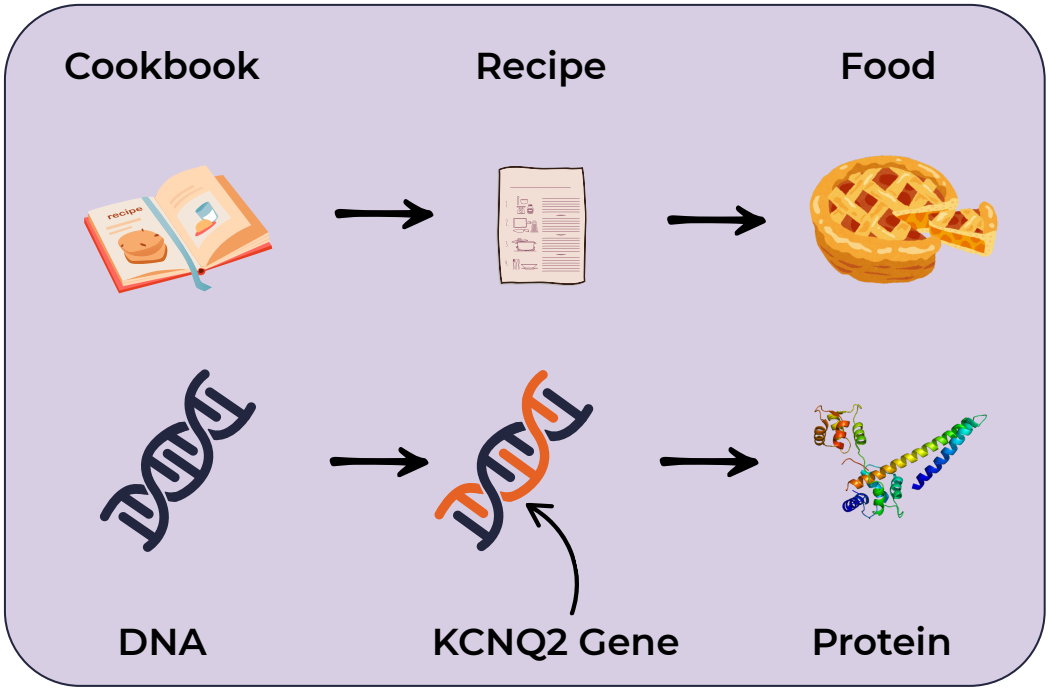
Each gene is a **recipe** that tells your body how to make a specific protein.

The **KCNQ2 gene** is one of these recipes. It provides instructions for making a protein called **Kv7.2**, which acts like a tiny **gatekeeper** in brain cells (neurons). These gates control the flow of potassium (K^+) — a key chemical that helps the brain's electrical signals stay balanced.

What Happens When the KCNQ2 Recipe Changes

Sometimes, a small change — or **typo** — occurs in this recipe. These changes are called genetic **mutations or variants.** Some are harmless, while others alter how the protein works or whether it's made at all.

When the KCNQ2 protein is affected, brain cells can become overactive or misfire, leading to seizures and developmental differences.



Every person's genetic "recipe" is unique — a mutation explains one part of your loved one's story but not the whole picture.

Types of Variants

There are several ways a gene's instructions can change. Here are some you might see on a genetic report:

- **Variant of Uncertain Significance (VUS)**: A change was found, but we don't yet know if it causes symptoms.
- **Missense Mutation**: One letter in the DNA code is swapped for another — like using salt instead of sugar in a recipe. The protein is still made but may not work as intended.
- **Insertions or Deletions** (Indels): Steps in the recipe are added or skipped, creating a scrambled or nonfunctional protein.
- **Duplications:** A DNA segment is copied one or more times, disrupting normal gene function.
- **Nonsense Mutation**: A "stop" signal appears too early, so the protein is cut short and often doesn't function.

Each of these changes can affect how potassium channels behave in the brain, which in turn influences how neurons communicate.

The gray cat ran down the hall.	**Original**
The gray cat ran down the **b**all	**Missense**
The gray **green** cat ran down the hall.	**Insertion**
The gray _____ ran down the hall.	**Deletion**
The gray **cat cat** ran down the hall.	**Duplication**
The gray**.**	**Nonsense**

Causal Variants

Every person has **two copies of the KCNQ2 gene** — one from each parent. Most children with KCNQ2-related disorders have a mutation in just one copy.

This means their body either:

- Doesn't make enough Kv7.2 protein, or
- Makes a version that doesn't work correctly.

Haploinsufficiency

In some cases, the body produces **less Kv7.2 protein** than it needs. It's as if the recipe is missing half the ingredients, so the brain ends up with fewer functioning potassium channels. With fewer channels to help control electrical signals, neurons can become **too excitable**. This is called **haploinsufficiency**, which means that one working copy of the gene isn't enough to keep things balanced.

Dominant Negative

In other cases, the change in the recipe doesn't just reduce how much protein is made — it causes the **altered proteins** to interfere with the **normal ones**. It's like baking a batch of cookies where one bad ingredient ruins the entire dough. These faulty Kv7.2 proteins can bind to healthy ones, preventing the channel from functioning correctly. This is known as a **dominant negative**, and it can cause more significant disruptions in how neurons send signals.

KCNQ2 proteins form part of potassium channels that help calm the brain's electrical activity. When these channels don't function properly, neurons can become **too excitable** — like a group of kids shouting over each other all at once.

This overactivity can cause seizures and affect how the brain develops early in life. Some mutations cause **loss-of-function**, while others cause **gain-of-function**. Both disrupt normal signaling — but in different ways.

Function Type

Before we dive in — yes, the alphabet soup continues: KCNQ2, K^+, LoF, GoF.
It's normal to feel overwhelmed by these terms. You're not alone — even doctors sometimes double-check the details.

This overactivity can cause **seizures** and affect how the brain develops early in life.
Some mutations cause **loss-of-function**, while others cause **gain-of-function**.
Both disrupt normal signaling — but in different ways.

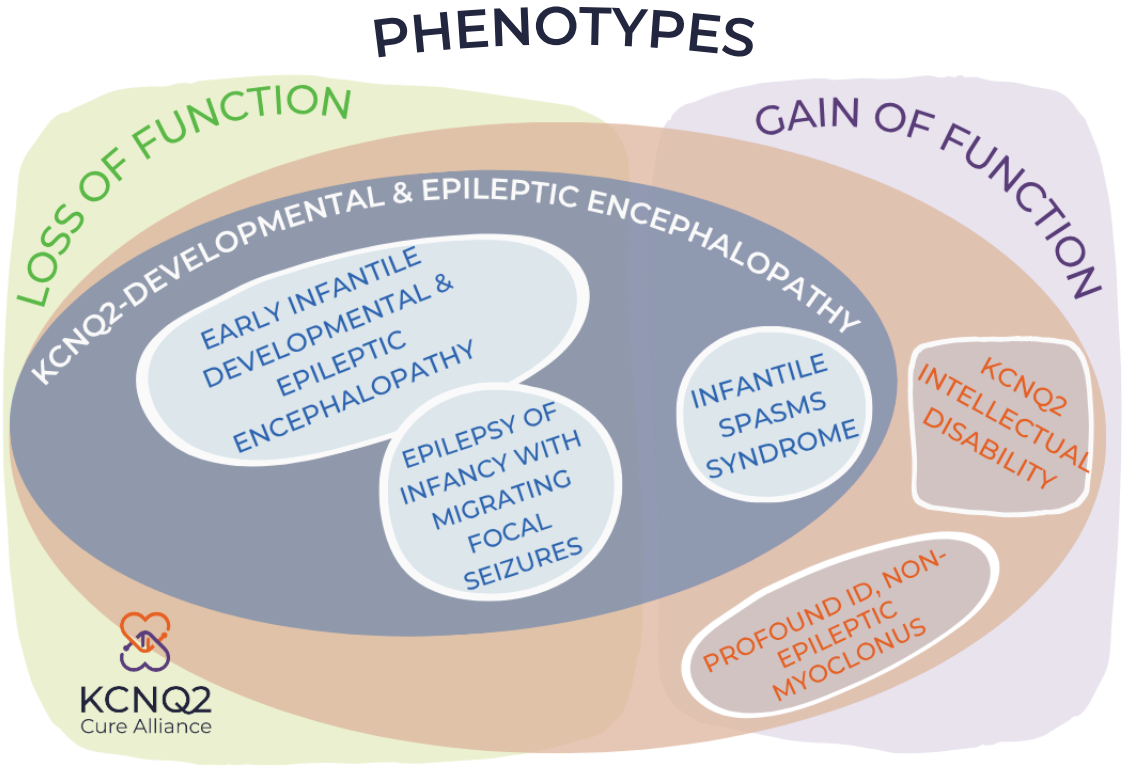
Here's what they mean in plain language:

Loss of Function (LoF)

- The potassium channel becomes too hard to open or doesn't open enough.
- Too little potassium exits the cell.
- Neurons become overly active and fire too easily.
- This overactivity can trigger seizures or disrupt normal brain signaling.

Gain of Function (GoF)

- The channel opens too easily or stays open too long.
- Too much potassium leaves the cell.
- The neuron becomes less responsive, slowing communication between brain cells.



Why Knowing Matters

Knowing whether a variant causes **loss-of-function** or **gain-of-function** can help guide treatment choices.

For example:

- **Sodium channel blockers** often help in loss-of-function cases.
- The same drugs might worsen symptoms in gain-of-function cases.

However, most clinical genetic reports **don't specify** whether a mutation is GoF or LoF.

This determination usually requires consultation with a geneticist or neurologist familiar with KCNQ2 — and sometimes additional laboratory or research testing.

Why Results Can Change Over Time

Genetic test results don't change — but our understanding of them does.
As research advances, doctors may learn new information about specific variants.

If your loved one's report is more than a few years old, ask your care team if it should be **re-analyzed.** Updated tools and databases may now provide answers that weren't available before.

Staying connected with your genetic provider and **KCNQ2 Cure Alliance** ensures you'll hear about new discoveries as they happen.

The goal isn't for you to become an expert in genetics — it's to help your care team understand how to choose the safest and most effective treatments.

Inherited vs. De Novo Mutations

Most KCNQ2 mutations happen **by chance**, not because of anything parents did or didn't do. These are called **de novo mutations**, meaning they occurred randomly in the egg, sperm, or very early in development.

In some cases, a mutation can be **inherited**, even if no one else in the family has symptoms.

This may happen through:

- **Mosaicism:** When only some of a parent's cells carry the mutation.
- **Germline mosaicism:** When the mutation is present only in reproductive cells (sperm or egg), allowing it to be passed on even though the parent shows no signs of the disorder.

If you're planning future pregnancies, a **genetic counselor** can explain recurrence risks and discuss options such as carrier testing or prenatal testing.

Genetic information can feel heavy — but it also empowers families to make informed choices and drive research forward.

Cambridge Elements

Want more than the basics? ***Cambridge's KCNQ2/KCNQ3 "Elements"*** guide covers the history of KCNQ2, deeper science for those who want it, and real stories from parents. It is written for both families and professionals, so parts may feel technical. Use what helps and skip what you do not need.

Understanding Your Loved One's Genetic Results

One of the most common questions families ask is:

"What does this result mean for my loved one's future?"

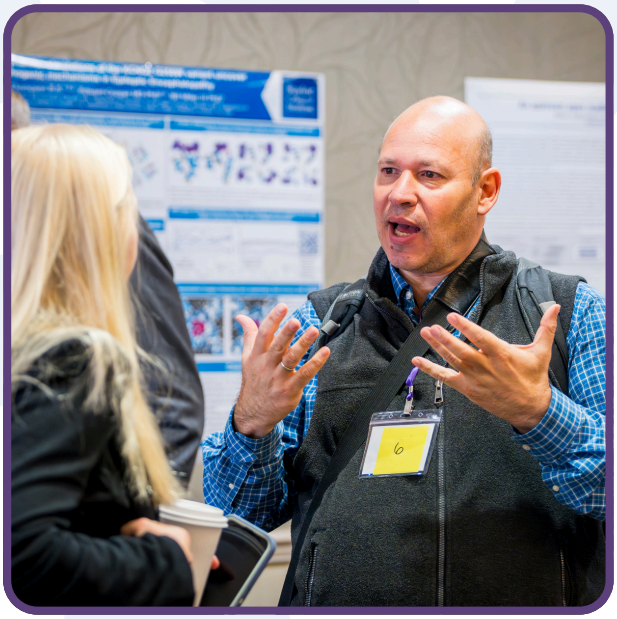
The short answer: **Genetic testing gives clues, not predictions.**

KCNQ2-related disorders exist on a wide spectrum. Even people with the same mutation can have very different experiences.

Here's why outcomes vary so widely:

1. **Different mutations** change the protein in different ways.
2. **Other genes** and environmental factors influence how the brain adapts.
3. **Early treatment and therapies** can shape long-term development.
4. **Science evolves.** What's uncertain today may be clear in a few years.

That's why your doctor looks at both your child's genetic report and their individual symptoms — not one in isolation.



From DNA to Discovery: How Families Advance KCNQ2 Research

KCNQ2 may be rare, but research is growing rapidly — and families are leading the way.

When you share your story, complete a survey, or participate in a study, you help scientists understand how different mutations impact real people.
Your child's experiences become part of the bigger picture, helping identify patterns and guide therapies.

1. Basic Science Research

This is the "behind-the-scenes" lab work that explores the biology of KCNQ2.
Scientists use:

- Animal models (like mice or zebrafish)
- Human cells grown in the lab
- Computer simulations

These studies ask:

- How do specific mutations change potassium channels?
- Why do seizures happen?
- Which drug compounds might help restore balance?

This work builds the foundation for all future treatments.

2. Clinical Research

This type of research involves families directly.
It includes:

- Surveys and medical record reviews
- Clinical trials
- Observational studies

Clinical research helps doctors understand:

- How KCNQ2 affects people over time
- What therapies and interventions make the most difference
- How to set better standards of care

Both types of research work together — one in the lab, one in real life — to drive progress faster.

How You Can Get Involved

You don't have to be a scientist to make a difference. Many research studies today can be completed right from home using a computer or phone — no travel or special equipment needed.

Families play an essential role in shaping the future of KCNQ2 research. By sharing your story, your child's medical history, or participating in studies, you help scientists see the bigger picture of how KCNQ2-related disorders affect development, treatment response, and quality of life.

We maintain an active list of current research opportunities on our website, including studies open to families around the world. These may involve surveys, interviews, data sharing, observational research, or clinical trials — each one helping researchers better understand KCNQ2 and develop more effective therapies.

Visit our research participation page to explore ongoing opportunities, learn what's involved, and find the studies that best fit your family's situation:

🔗 KCNQ2 Research Opportunities

Your story matters. Every form you fill out, every survey you complete, and every study you join helps scientists unlock new insights and hope for the next generation.

The Future of Genetic Medicine for KCNQ2

Research into **genetic medicine** is growing rapidly, offering new hope for families affected by KCNQ2-related disorders. These approaches aim to treat — and one day potentially correct — the underlying cause of the condition, rather than just managing symptoms.

What This Means for Families

Each study, data registry, and family story helps scientists understand which approaches might be safest and most effective.

By sharing information and participating in research when possible, families play a vital role in shaping the path toward precision treatments — and eventually, a cure.

Types of Genetic Medicine

Antisense Oligonucleotides (ASOs)

ASOs are short, synthetic strands of genetic material designed to attach to specific RNA messages inside cells. You can think of them as tiny "molecular switches" that can turn a gene's activity up or down.

For some conditions, ASOs help the body make more of a missing protein; in others, they can silence a gene that's producing a harmful one.

Gene Replacement Therapy

Gene replacement works by delivering a healthy copy of the gene to cells that need it. This is usually done using a harmless, modified virus, that acts like a genetic delivery vehicle to carry the working KCNQ2 instructions into brain cells.

The goal is for these cells to begin producing functional Kv7.2 proteins again, restoring balance to the brain's electrical activity.

Gene Editing

Gene editing goes one step further by aiming to repair the gene itself. Using precise tools like CRISPR, scientists can target the exact "typo" in the DNA code and attempt to correct it at its source.

You're Not Alone

KCNQ2 may be rare, but you are part of a growing, global community of families, caregivers, researchers, and advocates walking this journey together.

Connection is powerful. Sharing your story, asking questions, and showing up — even on the hardest days — creates understanding, drives research, and builds a network of care that reaches far beyond any single diagnosis.

Whether you're looking for information, emotional support, or ways to get involved, **KCNQ2 Cure Alliance** is here to help you feel connected and empowered.

KCNQ2 Cure Alliance Resources

Founded and led by parents, KCNQ2 Cure Alliance is dedicated to supporting families like yours and advancing research that changes lives. From hosting family summits to funding studies and building educational tools, we're here to walk beside you through every stage of this journey.

While this handbook includes some basic resources to help you get started, the most up-to-date and comprehensive list of materials, programs, and opportunities can be found on our website. There you'll discover detailed guides, therapy and care tools, links to current research, and ways to connect with other families and professionals who understand KCNQ2-related disorders. We regularly update our online resource library to ensure you have access to the newest information, support options, and community initiatives.

🔗 Visit **www.kcnq2cure.org/resources** to explore more.

How to Stay Connected

KCNQ2 Global Support Network for Caregivers
The private family group on Facebook is often the first place new parents connect. You can ask questions, share updates, and find comfort in others who truly understand what you're going through.

🔗 Join our parent network

Newsletter & Email Updates
Stay informed about research, family stories, and upcoming events by subscribing to our newsletter.

🔗 Sign up here

KCNQ2 Summit
The bi-annual KCNQ2 Summit brings together families, clinicians, and researchers for an unforgettable weekend of learning, connection, and collaboration. Whether you attend in person or follow along online, you'll leave feeling supported, inspired, and part of something bigger.

Family Story Submissions
Every family's voice helps raise awareness. Share your KCNQ2 journey — the ups, the challenges, and everything in between.
Your story might be featured on our website or social media to help others feel less alone.

🔗 Submit your story

Volunteer Opportunities
Whether you have five minutes or five hours, there's a way to make an impact.
Volunteers help with events, mentorship, community outreach, and fundraising.

🔗 Sign up to volunteer

Resources

Research
Families are at the heart of scientific discovery. Learn about current studies, clinical trials, and ways to share your child's medical data securely through Citizen or partner research groups.

🔗 Explore current research opportunities

Educational Resources
Access webinars, printable toolkits, and downloadable guides created for different stages of the journey — from newly diagnosed families to experienced advocates.

🔗 Visit our resource library

A Closing Note

Every family in the KCNQ2 community is part of a much larger story — one of resilience, science, and love.
By learning, connecting, and participating, you're helping change the future of this rare condition for everyone who follows.

You don't have to do it all at once. You don't have to do it alone.

Take a breath. Reach out when you need help.
And remember: **you are part of a global community that believes in progress, hope, and each other.**

💜 "Alone we can do so little; together we can do so much."
— Helen Keller