

KCNT1-Related Epilepsy

Acute Care Summary for Emergency & Inpatient Teams

Educational summary to support acute care. Not a treatment protocol.

What Is KCNT1?

KCNT1-related epilepsy is a rare genetic epilepsy associated with early-onset, treatment-resistant seizures and significant medical complexity. Most affected children have developmental impairment, and the majority are nonverbal.

Many clinicians encounter only one or two KCNT1 patients in their careers.

Why Parent/Caregiver Input Is Critical

For children with KCNT1, parents and caregivers are the most reliable source of clinical information.

Because most patients are nonverbal and have atypical neurologic exams, changes in condition are often identified first by caregivers rather than monitors, labs, EEG, or imaging.

Caregiver observations should be treated as essential clinical data.

Understanding Baseline vs. Escalation

Children with KCNT1 often live with symptoms that would be considered emergencies in other children. Over time, families learn what is *typical* for their child.

Escalation means a meaningful change from that baseline.

Caregivers are trained to recognize when symptoms are:

- different in quality
- more severe
- longer-lasting
- not responding as usual

When caregivers say "*this is not normal for my child*", this should prompt careful evaluation, even if objective findings are subtle.

KCNT1-Specific Red Flags

Breathing and autonomic changes

- apnea or breathing pauses
- perioral cyanosis
- sudden color change, bradycardia, or desaturation
- may occur with seizures, around seizures, or without clear ictal activity

Unexplained distress

- prolonged crying or agitation that is different from baseline
- distress not clearly correlated with seizures or post-ictal state

Gastrointestinal symptoms with neurologic change

- feeding intolerance, vomiting, or abdominal discomfort
- especially when associated with lethargy, distress, or color change

Bleeding, hemoptysis, or unexplained anemia

- some individuals with KCNT1 have been reported to have systemic-to-pulmonary collateral arteries
- bleeding may be the first sign

Sudden loss of function

- loss of swallow, movement, alertness, or responsiveness compared to baseline

Imaging and Monitoring Considerations

- Absence of electrographic seizure activity does **not** exclude clinically significant deterioration.
- Routine imaging may miss systemic-to-pulmonary collateral vasculature.
- **CT angiography** may be more informative than standard imaging when bleeding, anemia, or unexplained respiratory compromise is present.

Key Principle

Listen to the caregiver.

In KCNT1, caregiver-reported change from baseline is often the earliest and most accurate indicator that escalation is needed.

Provided by the KCNT1 Epilepsy Foundation

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