

Epilepsy in KIF1A Associated Neurological Disorder (KAND)

KAND is a progressive developmental and neurodegenerative disorder caused by mutations affecting KIF1A, a long-range neuronal motor protein.

Approximately half of KAND patients have reported seizures, and half of epileptic KAND patients had their first seizure before age 10.

Epilepsy in KAND

- Seizures: 42%
 - Grand mal (tonic/clonic): 17%
 - Petit mal (absence) 29%
 - Atonic drop: 9%
 - Infantile Spasms: 4%



Epilepsy types vary between KAND patients, but **Developmental/Epileptic Encephalopathy with Spike Wave Activation in Sleep (DEE-SWAS)**, aka **Continuous Slow Spikes and Waves during Sleep (CSWS)**, is commonly observed in KAND.

For more information visit KIF1A.ORG's [Epilepsy in KAND](#) page, and our [Clinical Conversation on Epilepsy](#) with Dr. Tristan Sands, co-lead of the KAND EEG study.

What should I bring to my appointment?

Video recordings of suspected seizures are one of the best tools families can provide their epileptologist; seizures are stressful events and self-report/interviews may miss crucial details. Seeing which aspects of a patient's behavior change can inform clinicians about what might be happening in the brain.

What should I ask for?

Overnight/24hr EEG: Because KAND-associated DEE-SWAS/CSWS causes abnormal electrical activity during sleep, it is recommended to request an EEG with recorded sleep, or ideally an overnight/24hr EEG. These longer recordings are more likely to detect abnormal activity, including epileptic spikes and seizures.

Raw files for KAND EEG study: When scheduling an EEG, you can request that your physician provide a raw EEG data file afterward. Our clinical researchers need these data files for the first-ever [KAND-specific EEG study](#) to find patterns within our community. To participate:

- Contact the office or department that will, or has, *performed your EEG and request the original EEG full dataset (all the squiggly lines)—not just the summary report. If you've had multiple EEGs, gather as many full recordings as possible.*
- Medical records cannot provide you the EEG tracings but can provide study reports and other documents.
- Mail EEG data flash drives or CDs to Boston Children's Hospital for researchers to evaluate. Include the patient's name, date of birth, and your contact information. Mail to:

**ATTN: KIF1A Study
HIM, Chung Lab
4 Blackfan St., 9th Floor Room 907
Boston, MA 02115**

Email Study Coordinators at ASCENDstudy@childrens.harvard.edu to inform the research team when you mail the data. If you'd like, you can Cc: impact@kif1a.org in that email so we can track participation as well.