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 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.