

ASCEND

(Advancing the Science, Care, and Exploration of Neurodevelopmental Disorders)

Information/Instructions

The natural history study of KIF1A collects real-world data from individuals affected by KAND in order to gain valuable insights into the presentation of the disease over time. The study obtains an entire medical history as well as information on the current condition and experiences through online questionnaires and medical records you provide. Investigating the condition at different ages over time provides important understanding on the evolution of KAND across the lifetime. The collected information will allow researchers and clinicians to work toward improving medical management guidelines and support, as well as inform future clinical trials.

To ensure the most accurate understanding of life with KAND and the challenges being faced, it is essential that everyone participate. The natural history study provides an opportunity to assure that your family's issues and experiences with KAND are represented while supporting the research working toward treatment. Collecting information from every affected family also ensures that all symptom-causing mutations are included in future research considerations. So, please join us and the KIF1A community in this step toward improving the lives of everyone affected by a KAND.

If you would like to participate in our natural history study of KIF1A, please read the following requirements and click the link below to get started.

The person signing the consent form for enrollment in the study and providing information must be able to sign legal and medical documents for the affected individual (e.g. guardian/healthcare-representative/proxy).

The person who completes the study surveys should be a guardian/caretaker who regularly observes and is intimately familiar with the affected individual through all aspects of their daily life, including their medical history, current health issues, emotions, behavior, sleep, diet, etc. This person should also be able to complete additional follow-up surveys in the future; it is important that the same person responds to future questionnaires to ensure consistent reporting.

Things you'll need:

- A copy of the full genetic testing report showing the exact mutation/diagnosis
- If you are a parent/relative/legal-guardian of an adult (over 18 years of age) and act as their medical proxy/healthcare-representative, you'll need a copy of a document showing that you are allowed to serve as their medical representative.

- If possible, a copy of any medical records that you have available for referencing to help answer questions

The variant of concern should be classified as Pathogenic, Likely Pathogenic, or Variant of Uncertain Significance. After filling out the intake form and uploading your genetic testing report, we will review the report and email you within 3 business days at the primary email address you provide on the intake form.

**If you are primarily interested in a clinical review of a variant or your experiences and are seeking clinical advice, you will need to schedule a clinical appointment for assessment of the genetic results and your case. A clinical appointment allows for a deeper review of the findings and possibly the ordering of additional testing to better serve the diagnosis. This also allows for clinical notes to formally be placed into the medical record.

If you've read all the above information and would like to participate in the natural history study of KIF1A, please click the following link to fill out the intake form and upload the necessary documents.

https://redcap.link/BCH_KIF1A

We will email you within 3 business days at the primary email address you provided on the intake form to move forward with your enrollment.

If issues or questions arise at any point during your participation in the study, please email ASCENDstudy@childrens.harvard.edu and we will reply as soon as possible.