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

Information / Instructions to be posted with entry link:

The natural history study of MAPK8IP3 collects real-world data from individuals affected by MAPK8IP3-related disorder in order to gain valuable insights into the presentation of the disease over time. The study obtains an entire medical history as well as the current condition and experiences through questionnaires and medical records. Investigating the condition at different ages over time provides important information on the evolution of MAPK8IP3-related disorder 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 MAPK8IP3-related disorder and the challenges being faced, it is essential that everyone participates. For affected-families, the natural history study provides an opportunity to assure your representation while supporting the research as we work toward treatments. So, please join us and the MAPK8IP3 community in this step toward improving the lives of everyone affected by a MAPK8IP3-related disorder.

If you would like to participate in our natural history study of MAPK8IP3, 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 legal/medical representative.
- If possible, a copy of any medical records that you have available for referencing to help answer questions

If you have a Variant of Uncertain Significance (VUS) and are interested in participating in our research study, please click the link, fill out the initial intake form and upload your report. We will review your report and reach out within 3 business days.

****If you are primarily interested in a clinical review of a variant/case 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 MAPK8IP3, please click the following link to fill out the intake form and upload the necessary documents.

https://redcap.sac-cu.org/surveys/?s=79MRYHT43PA8WK4K

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

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