

Study for caregivers of children with *STXBP1*-related disorder

The Children's Hospital of Philadelphia is looking for caretakers of children with *STXBP1*-related disorder to take part in an interview research study.

This interview research study will investigate (1) symptoms of *STXBP1*-related disorder; and (2) the impact of *STXBP1*-related disorder symptoms on the child and the caregiver.

This study seeks to better understand the current needs of *STXBP1*-related disorder families. The results of this study will help guide planning for future treatment options and clinical trial studies.

Who can participate?

- Caregivers of an individual with a confirmed diagnosis (molecular diagnosis, genetic diagnosis) of *STXBP1*-related disorder
- Caregivers must be over the age of 18 years old
- Caregivers may be biological parents, foster parents, grandparents, or other guardians that perform at least 50% of daily caretaking for a child with *STXBP1*-related disorder
- Eligible caregivers may have a child with *STXBP1*-related disorder of any age (Example: 6-month-old child with *STXBP1*-related disorder, 25-year-old child with *STXBP1*-related disorder)

Participants will be asked to:

- Consent to an audio recorded interview
- Complete a brief short-answer intake questions (Example: what is your age, what is your ethnicity, when was your child diagnosed with *STXBP1*-related disorder)
- Perform a 1 – 1 ½ hour phone or video-conference interview
- Discuss *STXBP1*-related disorder symptoms and the impacts of these symptoms on their child, themselves, and their family

Why participate?

- An opportunity to share your child's and your family's *STXBP1*-related disorder story
- Interview studies are necessary first steps towards clinical trial studies for rare disease communities
- Interview studies give insight into areas of health care management where improvement is needed for children and families

How to participate

- Contact ENGIN@chop.edu or 267-425-0515

Study for healthcare professionals involved in the care of individuals with *STXBP1*-related disorder

The Children's Hospital of Philadelphia is looking for healthcare professionals involved in the care of individuals with *STXBP1*-related disorder to take part in an interview research study.

This interview research study will investigate (1) symptoms of *STXBP1*-related disorder; and (2) the impact of *STXBP1*-related disorder symptoms on the child and the caregiver.

This study seeks to better understand the current needs of *STXBP1*-related disorder families and determine what symptoms and symptom-impacts are the highest priority for evaluation in future clinical studies. We believe that caregivers and healthcare professionals will describe similar but distinct *STXBP1*-related symptoms and symptom-impacts, motivating us to interview both caregivers and clinical experts.

Who can participate?

- Licensed physicians, nurses, genetic counselors, physical therapists, occupational therapists, speech therapists or other healthcare professional that manages that care of individuals with *STXBP1*-related disorder
- Healthcare professionals that evaluate at least 5 individuals with *STXBP1*-related disorder per year or manage the care of at least 3 individuals with *STXBP1*-related disorder
- Healthcare professionals that manage the care of individuals with *STXBP1*-related disorder in the pediatric or adult setting

Participants will be asked to:

- Complete a brief demographic survey
- Consent to an audio recorded interview
- Perform a 1 hour phone or video-conference interview
- Discuss *STXBP1*-related disorder symptoms and the impacts of these symptoms on their patients and their families

Why participate?

- Interview studies are necessary first steps towards clinical trial studies for rare disease communities
- Opportunity to support a rare disease community in your specialty

How to participate

- Contact ENGIN@chop.edu or 267-425-0515