STXBP1 and SYNGAP1 Related Disorders Natural History Study

NCT06555965

Status	RECRUITING
Sponsor	Children's Hospital of Philadelphia
Enrollment	600 participants

Key Eligibility Criteria

Inclusion (2)

- Male or female of any age.
- Presence of a STXBP1 or SYNGAP1 gene mutation. The variant in STXBP1 or SYNGAP1 must be classified as causative based on clinical and variant classification criteria. Historical documentation is sufficient to support eligibility for the study. Confirmatory testing will be obtained, if necessary, at baseline and performed by a CLIA certified laboratory.

Exclusion (5)

- The presence of a confirmed mutation in a gene other than STXBP1 or SYNGAP1 that is known to contribute to a neurodevelopmental disability. This includes full gene deletions of STXBP1 or SYNGAP1 that include other genes beyond STXBP1 or SYNGAP1.
- The presence of a significant non-STXBP1-RD or non-SYNGAP1-RD related central nervous impairment/behavioral disturbance that would confound the scientific rigor or interpretation of results of the study.
- History of intraventricular hemorrhage, structural brain deficit or congenital heart disease
- The presence of a clinical comorbidity deemed by the investigator to potentially confound the typical presentation of STXBP1-RD or SYNGAP1-RD.
- Pregnant women or females of age of menarche who are found to be pregnant upon urine pregnancy testing.

Locations (5 total)

Stanford Medicine Children's Health, Palo Alto, California, United States
Children's Hospital Colorado, Aurora, Colorado, United States
Weill Cornell Medicine, New York, New York, United States
... and 2 more locations