A Natural History Study of Angelman Syndrome

NCT07417137

Status	RECRUITING
Sponsor	Massachusetts General Hospital
Enrollment	40 participants

Key Eligibility Criteria

Inclusion (10)

- The participant has a primary clinical diagnosis of Angelman syndrome with documented genetic variation(s) affecting the function of the UBE3A gene within the human 15q11.2-q13.3 locus. Co-occurring conditions (e.g., autism spectrum disorder, cerebral palsy, intellectual disability) are permitted; however, Angelman syndrome must be the primary clinical diagnosis.
- The participant is male or female (assigned sex at birth) and aged e1 year at the initial study visit.
- The participant has a study partner who meets the study partner criteria below.
- The participant, if unable to provide informed consent, has an appropriate surrogate who is at least 18 years of age and willing and able to provide informed consent on behalf of the participant in accordance with current International Council for Harmonisation (ICH) guidelines and applicable institutional regulations.
- Individuals must satisfy the following criteria to be enrolled as study partners:

... and 5 more (see full listing online)

Exclusion (10)

- The participant has at least one additional known genetic abnormality outside the human 15q11.2-q13.3 locus causing a probable or known developmental disability.
- At least one standard-of-care treatment (medication or adjunctive therapy) used by the participant was changed during the 28 days (4 weeks) prior to the first study visit. Treatments include, but are not limited to, doses of anti-epileptic medications, behavioral management medications, sleep medications, gabapentin, cannabidiol, special diets, supplements, speech therapy, occupational therapy, applied behavioral analysis (ABA), psychosocial interventions, physical therapy, or nutritional support.
- The participant has unstable epilepsy, defined as having an emergency department visit or hospitalization for seizure-related concerns within the 28 days (4 weeks) preceding the initial study visit.
- The participant is of childbearing potential and is either pregnant, breastfeeding, or not using an adequate method of contraception; abstinence is acceptable.
- The participant has a clinically relevant history of malignancy; clinically significant abnormal test results; clinically significant cardiovascular, hematologic, hepatic, muscular, neurologic, or renal disease; or has experienced other clinical events which, in the opinion of the investigator, render participation unsuitable.

... and 5 more (see full listing online)

Locations (1 total)

MGH Lurie Center for Autism, Lexington, Massachusetts, United States