Research into potential therapies for Angelman Syndrome (AS) is advancing, with increasing opportunities for clinical trials in the future. There are currently no disease modifying therapies available for AS, and care is limited to supportive interventions attempting to address behavioural, motor, communication and sleep needs, combined with symptomatic treatment of medical complications, including seizures and sleep deficits.

There is a need to develop innovative endpoints for AS, leveraging the availability of digital and decentralised (home based) strategies to reduce burden of participation and increase value for families, and focusing on specific aspects of AS such as electrophysiology and sleep. These measures could be informative biomarkers or meaningful endpoints for future drug studies.

This is a non-drug study to investigate novel endpoints in AS. It is a prospective longitudinal study conducted at several clinical centers in the US including individuals with AS and volunteers, designed to deeply phenotype a small number of individuals over a period of 12 months per individual. The focus will be to characterise sleep features, electrophysiology, and neurodevelopment in individuals with AS, combining clinic visits with home based assessments and digital devices. The acquisition of gold standard measures such as polysomnography (PSG) done in the home setting will be compared with data acquired from digital devices including a sleep mat and digital watch. Additional assessments and questionnaires in the home and clinic will evaluate neurodevelopment, adaptive skills, communication and quality of life. The goal is to develop innovative and feasible biomarkers and clinical endpoints for future treatment studies in AS.