

Global Angelman Syndrome Registry

Registry Data Catalogue

About this catalogue

This catalogue provides a high-level overview of the information currently collected by the Global Angelman Syndrome Registry. It is intended to help researchers, clinicians and collaborators understand the scope of data available within the Registry.

The Registry combines core demographic and clinical information with longitudinal follow-up data and standardised outcome measures to support natural history studies, clinical research and health technology assessment (HTA).

This catalogue is intended as a high-level reference and does not list every individual data element, response option or variable collected. Many categories contain multiple related data fields. Researchers requiring more detailed information are encouraged to contact the Registry team.

Collection schedules vary across the Registry. Some information is collected once (for example, diagnosis and genotype), while other information is updated over time to capture changes throughout the lifespan.

Note: The Registry was redesigned in 2026. Some historical data are identified as archived within this catalogue. Historical data remains available for approved research. *Read more about the Registry redesign.*

Current Data Collection

Demographics

Collection: Baseline (updated as required)

- Participant demographics
- Country and location
- Family information
- Diagnosis details
- Genetic subtype

History of Diagnosis

Collection: Baseline

- Diagnosis history and pathway
- Genetic testing and diagnosis confirmation

Clinical

Collection: Longitudinal

Medical conditions

- Growth and feeding

- Eyes and vision
- Brain and nervous system (including epilepsy)
- Behavioural and psychiatric conditions

Clinical-Medical conditions (cont)

- Musculoskeletal conditions
- Respiratory conditions
- Digestive system conditions

Hospitalisations and procedures

- Hospitalisations
- Surgical history
- Anaesthesia

Medications

- Current medications
- Medication changes

Therapies

- Therapy participation
- Therapy changes

Clinical trials

- Clinical trial participation

Specialist care

- Specialty clinic attendance

Development

Collection: Longitudinal

- Gross and fine motor abilities
- Activities of daily living
- Continence
- Favoured activities and interests

Sleep

Collection: Longitudinal

- General sleep questions
- Sleep Disturbance Scale (SDS)

Standardised Assessments

Collection: Scheduled intervals

- Aberrant Behaviour Checklist – Community (ABC-C)
- Observer-Reported Communication Ability (ORCA)

Archived Data

The following information was collected in previous versions of the Registry and remains available for approved research purposes. These data are no longer routinely updated.

Detailed Epilepsy

- General seizure history
- Atonic seizures
- Tonic, clonic and tonic-clonic seizures
- Myoclonic seizures
- Cluster motor seizures
- Absence seizures
- Cluster non-motor seizures
- Epileptic spasms
- Convulsive status epilepticus
- Non-convulsive status epilepticus
- Unknown or unclassified seizure types

Newborn & Infancy History

- Early living arrangements
- Infant feeding
- Temperature regulation
- Behavioural and developmental concerns
- Respiratory issues
- Other early medical conditions

Communication

- Communication methods
- Augmentative and Alternative Communication (AAC)

Pathology & Diagnostics

- Additional diagnostic testing

Sleep Diary

- Historical sleep diary records

Data Access

Researchers interested in accessing de-identified Registry data are encouraged to visit the **Request Data Access** section of the Registry website for information on eligibility, governance requirements and the application process.

Queries? Contact curator@angelmanregistry.info