Angelman syndrome (AS) is a rare, genetic, neurodevelopmental condition characterized by heterogeneous symptomatology, including behavior-related abnormalities, motor and communication impairment, and sleep disturbances. Currently, available treatments address only symptoms without the ability to target AS-specific pathophysiology. Heterogeneity of symptomology in AS poses a challenge to the selection of relevant measures of efficacy for therapeutic clinical trials. Throughout appreciation of the aspects of AS that are most salient and challenging for patients and their caregivers is essential to successful development and evaluation of new treatments. One group discussion and 6 individual interviews with caregiver-parents of pediatric and adult patients with AS aged 2–28 years were conducted to gather information on challenges of care.

Caregiver-Parent Domain Rankings

Objectives
- Caregivers are parents who care for patients with Angelman syndrome (AS). Their role is to support and enhance the patient's development, including the areas of communication, behavior, anxiety behavior, motor, and sleep.
- The domains are considered based on the challenges experienced by caregivers, with each domain having specific characteristics.

Background
- Angelman syndrome (AS) is a rare, genetic, neurodevelopmental condition characterized by heterogeneous symptomatology, including behavior-related abnormalities, motor and communication impairment, and sleep disturbances. Currently, available treatments address only symptoms without the ability to target AS-specific pathophysiology.
- Heterogeneity of symptomology in AS poses a challenge to the selection of relevant measures of efficacy for therapeutic clinical trials.
- Throughout appreciation of the aspects of AS that are most salient and challenging for patients and their caregivers is essential to successful development and evaluation of new treatments.

Methodology
- One group discussion and 6 individual interviews with caregiver-parents of pediatric and adult patients with AS aged 2–28 years were conducted to gather information on challenges of care.

Domains in Depth – Communication

- Communication: Caregivers report the following about patients’ communication (Table 1):
  - Many patients do not speak or express themselves verbally.
  - Some patients can communicate using sign language or picture communication systems.
  - Patients may overuse and mispronounce words.
  - Eye gaze is a primary tool for communication.
  - AAC devices are used to augment communication.
  - Patients may express their needs and wants through touch, physical restraint, and inappropriate behaviors.

Domains in Depth – Behavior

- Behavior: Caregivers report the following about patients’ behavior (Table 3):
  - Patients may have limited ability to engage in age-appropriate behaviors.
  - Patients may engage in repetitive behaviors.
  - Patients may have difficulties with attention and distractibility.
  - Patients may have problems with aggression and irritability.
  - Patients may have difficulties with feeding and eating.

Domains in Depth – Anxiety

- Anxiety: Caregivers report the following about patients’ anxiety (Table 4):
  - Patients may have significant anxiety about sleep, social situations, and new experiences.
  - Patients may have difficulties with separation anxiety.
  - Patients may have fears about loud noises and sudden changes in the environment.

Domains in Depth – Sleep

- Sleep: Caregivers report the following about patients’ sleep (Table 5):
  - Patients may have difficulties with falling asleep, staying asleep, or waking up multiple times during the night.
  - Patients may have difficulties with sleep patterns due to circadian rhythm disturbance.
  - Patients may have difficulties with daytime sleepiness.

Results
- Caregivers indicated that communication, behavior, anxiety behavior, motor, and sleep are the most significant challenges they face.
- Research is needed to develop better interventions that address these challenges.

Conclusions
- Further research is needed to develop better interventions for these domains.
- Caregiver education and support is essential for improving patient outcomes.