

ANGELMAN SYNDROME

Voice of the Patient Report



The Angelman syndrome (AS) community had the unique opportunity to share our experiences on living with the condition and perspectives on treatments for our loved ones directly with the Food and Drug Administration (FDA). On April 7, 2025, the Foundation for Angelman Syndrome Therapeutics and the Angelman Syndrome Foundation cohosted a virtual externally-led patient-focused drug development (EL-PFDD) meeting including nearly 400 participants:

227 AS caregivers and family members

41 FDA staff

67 drug developer company representatives

27 researchers and health care providers

This “**Hope in Action: an EL-PFDD Meeting on Angelman Syndrome**” and the resulting Voice of the Patient Report, submitted to FDA in November 2025, created a platform for our community to share insights and priorities directly with the FDA and other stakeholders, ultimately shaping the future of therapy development.

Angelman syndrome is a serious neurogenetic condition that affects the brain and causes profound developmental and cognitive challenges across the typical lifespan. Current treatments only address symptoms and have minimal effects on how individuals with AS feel or function. The EL-PFDD Meeting highlighted the need for increased awareness of the impact of AS, on both the person living with the condition and their caregivers, as well as the need for improved treatments.

Key Takeaways:

- The symptoms of AS are wide-ranging and complex, with devastating impacts on an individual’s activities of daily living that significantly limit his or her independence.
- **Communication challenges, cognitive limitations, challenging behaviors, and sleep disturbances** are among the symptoms that place the greatest strain on people living with AS and their caregivers.
- Treatment of AS is limited to symptom management through a dizzying array of medications, assistive technologies, and other interventions deployed across a normal lifespan with minimal improvements in quality of life at best.
- AS has significant physical and mental impacts on both the individuals affected and their caregivers. **Caregivers must provide constant care for the entirety of their child’s life.**
- There is an urgent need for a disease-modifying treatment for AS. Families are eager for a treatment that could provide **even small improvements** in cognition, communication, and the ability of their loved ones to carry out activities of daily living.
- The need for FDA concurrence on the utility of existing outcome measures and possible biomarkers intended to measure potential effects of new disease-modifying treatments will help advance late-stage trials. Such advances could accelerate innovation to address AS unmet needs.

“When it comes to those challenges, I would say nonverbal communication is number one. I feel like the behaviors are a result of him not being able to communicate quickly and verbally is something that, if he had five words, I would give my life for him to have five words.”

– *Mother of nine-year-old son*

“Our son requires care in everything he does, helping him with dressing, toileting, and getting to school. Independence is non-existent, which is unusual for an 11-year-old. As he gets older and bigger, so do his needs.”

– *Mother of 11-year-old son*

“As an adult, he’s had the experience that I have most feared. He’s big, he looks like an adult when he doesn’t feel like getting shaved. And he had a caregiver in his group home who wasn’t familiar with him, who panicked because of his behaviors and called the police who then came into the home, and wanted to take him to the emergency room...”

– *Mother of 23-year-old son*

“[Our son] never slept through the night, which means that he must be awake or one of us must be awake with him 24/7. So my husband and I do take turns caring for him try and prevent caregiver burnout.”

– *Mother of nine-year-old son*

Scan the QR code to read the Angelman Syndrome Voice of the Patient report

