Provider Guide

How to Use this Toolkit for Patients with Angelman Syndrome

Thank you for your commitment to supporting individuals with Angelman syndrome. Your willingness to learn, listen, and provide compassionate care makes a profound difference in the lives of your patients and their families.

If a caregiver has shared the Angelman Syndrome Foundation (ASF) Transition of Care Toolkit with you, it likely means you're working with someone who is not only deeply invested in their loved one's wellbeing but also eager to partner with you. Caregivers of individuals with Angelman syndrome often serve as the primary coordinators of care, advocates, historians, and communicators for their loved ones. By choosing to engage with this resource, you're taking an important step in building a collaborative, person-centered care plan—one that honors the caregiver's knowledge and the patient's unique needs.

The transition of care from pediatric to adult-based healthcare is a critical phase for patients with Angelman syndrome, their families, caregivers, and providers. The Provider Guide is designed to support healthcare providers in how to use the toolkit effectively to ensure a smooth, informed, and well-coordinated transition. The toolkit centralizes essential information and serves as a resource to streamline communication and continuity of care.

Purpose and Scope

This toolkit is designed to be a comprehensive resource managed by the caregiver with the support of their providers. It has been created to help guide both parties through the complexities of transitioning from pediatric to adult care by centralizing crucial data including:

- An extensive review of the patient's medical history including all diagnoses, medications, allergies, and past surgeries or procedures
- Emergency care plans
- Past and current provider details

How Providers Should Use the Toolkit

Before Transition	• Use the toolkit to review important patient information and specific needs related to the care of the patient. • Engage in family and caregiver education. As a provider, it is essential to ensure that the family, caregivers, and the patient (as much as possible) are involved in the transition process. The toolkit includes sections designed to help caregivers understand what to expect during the transition, how to manage ongoing care, and when to reach out for specialized help.

During Transition	• Pediatric providers, use this toolkit as a prompt to help caregivers create a centralized document to pass medical information to the adult care team. It should be shared by caregivers with their loved ones' new adult providers to help them understand the patient's unique needs and ongoing treatments or therapies which need to be continued and/or adjusted. • Help caregivers keep care plans updated and reflective of adult-specific needs by reviewing and documenting necessary information (e.g. medication adjustments, changes in care protocols, or a specialized approach to symptoms management in adults with AS).
After Transition	• Attempt to maintain clear and open channels of communication between the pediatric and adult care teams. The toolkit is designed to help maintain continuity of care and facilitate ongoing discussions about the patient's evolving needs. • Prompt caregivers to update toolkit with changes in the patient's medical condition, new treatments or emerging symptoms in order to maintain a current record that both pediatric and adult teams can rely on as the patient's care evolves.

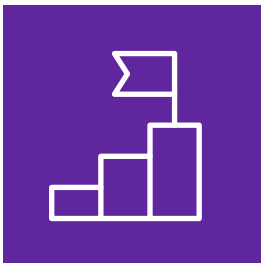
Successful transition requires collaboration among a team of healthcare providers. Providers should encourage dialogue amongst all involved aiming to address all aspects of the patient's healthcare. The toolkit is not a stand-alone solution, but a collaborative tool that should be used in conjunction with discussion among:

- Pediatric specialists
- Adult providers
- The patient and his/her caregivers

Advice Directly from Angelman Syndrome Expert Providers

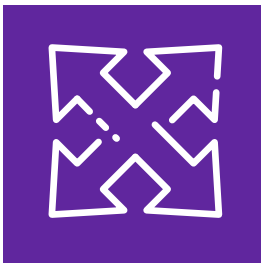
Avoid Diagnostic Overshadowing: A diagnosis of Angelman syndrome does not automatically explain new or changing symptoms. Common medical issues should always be ruled out first. If a patient with AS presents with changes in behavior such as increased agitation, social withdrawal, or increased drowsiness consider underlying illness, pain, or discomfort and evaluate as appropriate. Behavioral shifts are often signs of medical problems that the patient may be unable to verbally express.

Familiarize Yourself with Common Medical and Developmental Features of AS: Individuals with AS often experience seizures (which may lessen with age), sleep disturbances, gastrointestinal issues, gait abnormalities and ataxia, and limited or nonverbal communication. A consistently happy demeanor is characteristic but should not be mistaken for an absence of distress. It is important to distinguish typical AS behaviors, such as frequent laughter or hand-flapping, from signs that may indicate pain or discomfort.



Monitor Changes Over Time: Avoid assumptions that the patient's current state is his/her permanent baseline. Individuals with AS can continue to make gains with appropriate support. Further, they may decline if unrecognized medical needs go unmet. Encourage caregivers to track changes in seizure activity, motor skills, mobility, sleep, GI function, feeding tolerance, and communication efforts over time . Longitudinal monitoring and caregiver input are essential for determining whether or not interventions are beneficial.

Engage the Patient at Every Encounter: People with AS often have strong receptive language skills. Always assume competence. Speak directly to the patient rather than talking at, through, or around the patient, or only speaking with the caregiver. Always explain what you're doing even if the patient does not respond verbally. If he/she uses an AAC device (e.g., tablet or communication board), ask how it works and allow extra time for responses.



Be Adaptable: To the best of your ability, create a supportive, sensory-considerate setting. Schedule longer visits when possible, avoid unnecessary physical handling or restraint, and minimize triggers such as harsh lighting, crowded or noisy waiting areas, and long delays. Be flexible. For instance, care may be more effective if the patient is seated with a caregiver or even on the floor.

Foster Collaborative, Lifelong Care: Caring for an individual with AS requires ongoing partnership. It is important to trust the caregivers' insight; they are your most valuable diagnostic ally and are often the first to notice subtle changes in behavior, mood, appetite, or sleep that may indicate an underlying issue even when exams and labs appear normal. Collaborate with specialists, support access to AAC, durable medical equipment, therapies, and community resources. Emphasize proactive wellness care—not just crisis management—to help your patient thrive.

Stay Curious! Angelman syndrome is rare and complex, but families value providers who take the time to ask questions, seek resources, and remain open to learning. A curious, informed approach builds trust and leads to better care.

Advice from Caregivers: What We Wish Providers Knew

Caregivers are experts in their loved one's care—and they want to be treated as respected partners on the healthcare team. Here's what they want you to know:

- **We know our child best.** If something seems off—even subtly—we often notice it first. Please take our instincts to heart, even if tests or vitals seem normal.
- **We want to work with you.** At home, we're juggling medications, seizures, feeding schedules, insurance paperwork, adaptive equipment, and more. We've had to become experts in Angelman syndrome, and we're here to collaborate—not to control.
- **We want to build a relationship, not just get through an appointment.** Compassion, flexibility, and curiosity from providers go a long way. When you speak directly to our loved one, ask questions, and show a willingness to learn, it builds trust.
- **We're in this for the long haul.** This isn't just a transition—it's a part of our journey. When you show you're in it with us, it gives us strength and hope.

The transition of care for patients with Angelman syndrome requires thoughtful planning, clear communication, and an ongoing commitment to high-quality care. By using this toolkit, healthcare providers can help ensure that the patient's journey from pediatric to adult care is as seamless and supportive as possible. This guide should be used alongside the toolkit to help healthcare providers navigate the process with confidence, providing the best care for the patient while addressing the needs of their families and caregivers.

Clinician Resources & Support

- **Training and Education:** Various organizations including the Angelman Syndrome Foundation and the Child Neurology Foundation offer related educational resources. Providers should take advantage of training opportunities related to Angelman syndrome to stay up-to-date with the latest research and best practices in care by subscribing to relevant newsletters and becoming familiar with the resources available.
- **Emergency Care Planning:** One of the critical components of this toolkit is ensuring providers are aware of emergency care plans. These should be reviewed regularly to ensure they are appropriate for the patient as he/she ages and care needs change.

- **Current Best Practices:** The Angelman Syndrome Clinical Care Toolkit has been developed in close collaboration with leading medical professionals. It is based on the most current scientific evidence and clinical experience, providing a trusted resource that can support and empower healthcare providers in their care of individuals with Angelman syndrome. Included are:

  - **Standards of Care**
    - A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome (Duis et al, 2022) PMID: 35150089

  - **Epilepsy Management**
    - Low glycemic index treatment for seizures in Angelman syndrome (Thibert et al, 2012) PMID: 22779920
    - Myoclonus in Angelman syndrome (Pollack et al, 2017) PMID: 29555100
    - Seizure treatment in Angelman syndrome: A case series from the Angelman Syndrome Clinic at Massachusetts General Hospital (Shaaya et al, 2016) PMID: 27206232

  - **Additional Resources**
    - Clinical Guidance for the Treatment of Seizures in Angelman Syndrome
    - Management of Non-convulsive Status Epilepticus in Angelman Syndrome
    - Preparing Your Individual with Angelman Syndrome for General Anesthesia
    - Aspiration Prevention in Angelman Syndrome
    - General Anesthesia Best Practices
    - Angelman Syndrome Foundation Resources: Videos, podcast episodes, educational content, group support, and more