

Case Study: Angelman Syndrome (AS)

Part 1: Getting to Know Devon

Devon and His Family

Devon is a 30-month-old boy who lives in a suburban neighborhood in Middle America with his mother, Katherine (32), father, Greg (33), and older sister, Lucy (5). The family lives near lots of Katherine's relatives as she grew up in the town as she is now raising her family. Katherine works part-time as a dental office receptionist, and Greg is a certified HVAC technician employed by a regional company. The family is considered middle-class and lives in a modest single-family home. They have a supportive relationship with their pediatrician and are actively involved in their community, including attending events at Lucy's elementary school and participating in neighborhood activities.

Devon has always been a joyful child, frequently smiling and laughing. However, his parents began noticing developmental delays around 12 months, including limited speech, difficulty sitting independently, and frequent jerky movements. Devon enjoys music and brightly colored toys but struggles with balance and coordination. He often flaps his hands when excited and has trouble sleeping through the night.

Medical and Developmental History

Devon was born full-term with no complications. His early infancy was marked by feeding difficulties and hypotonia. At 18 months, Katherine and Greg expressed concerns to their pediatrician about Devon's delayed milestones and lack of verbal communication. A referral was made for genetic testing, which confirmed a diagnosis of Angelman Syndrome due to a deletion on the maternal chromosome 15q11–q13 region.

The family was unfamiliar with AS and felt overwhelmed by the diagnosis. They were connected with a genetic counselor and early intervention services. Katherine and Greg expressed a desire to learn how to support Devon's development while maintaining their cultural values and routines.

Discussion Prompts:

- What additional questions might you ask to better understand Devon's family context and support system?
- What might you want to know more about if you were on Devon's team?
- What strategies would you use to develop a strong partnership with his family? and rural environment, what strategies could help him engage more comfortably in community settings?

Part 2: Screening and Assessment

Devon's journey toward diagnosis began when his parents, Katherine and Greg, noticed that he wasn't meeting developmental milestones typical for his age. By 18 months, Devon had not yet spoken any words, struggled to sit independently, and exhibited frequent, jerky movements. Although he was a cheerful child who smiled and laughed often, his motor coordination and communication delays raised concerns.

During a routine pediatric visit, Katherine shared her worries with their trusted pediatrician, who recommended a referral to a developmental specialist. The initial evaluation included comprehensive developmental screening and clinical observation. The specialist noted hallmark features of Angelman Syndrome (AS): frequent laughter, minimal speech, ataxia (uncoordinated movements), and global developmental delays. Devon's joyful demeanor stood out, but so did his difficulty with balance, he often stumbled or fell during play, and his inability to use words to communicate.

The specialist ordered an EEG, which revealed a characteristic pattern often seen in children with AS. This prompted further genetic testing, including methylation analysis and DNA sequencing. The waiting period for results was emotionally taxing for Katherine and Greg, who spent hours researching possible outcomes and trying to prepare themselves for what might come. When the results confirmed a deletion on the maternal chromosome 15q11–q13 region, consistent with Angelman Syndrome, the family felt a mix of relief at having an answer, and grief over the uncertainty of what the future might hold.

Following the diagnosis, Devon was referred to a multidisciplinary team for a full evaluation. This team included a developmental pediatrician, neurologist, genetic counselor, speech-language pathologist, occupational therapist, and physical therapist. The team conducted both clinic-based assessments and home visits to gain a holistic understanding of Devon's functioning.

During home visits, Devon was observed to be highly social, often seeking eye contact and physical closeness with family members. He used gestures, facial expressions, and vocalizations to communicate his needs and emotions, but he still had no spoken words. His play was joyful but uncoordinated; he frequently lost his balance and had difficulty navigating his environment. Katherine and Greg also reported persistent sleep disturbances—Devon had trouble falling asleep and often woke multiple times during the night, leaving the entire family exhausted.

The evaluation team took time to explain the diagnosis and its implications, using visual aids and culturally sensitive language to ensure Katherine and Greg felt informed and supported. The family was encouraged to ask questions and share their concerns, which ranged from

communication strategies to long-term educational planning. The team emphasized Devon's strengths—his social engagement, joyful personality, and responsiveness to music and visual stimuli—and reassured the family that early intervention could make a meaningful difference.

Discussion Prompts:

- What challenges might arise when distinguishing AS from other developmental disorders?
- How can providers support families in understanding genetic testing results?
- What are the benefits of observing children in both clinical and home settings?

Part 3: Navigating Nursing Care in Preschool

When Devon was accepted into a local inclusive preschool program, Katherine and Greg were both excited and anxious. While they were thrilled about the opportunity for Devon to engage with peers and participate in structured learning, they were deeply concerned about how his seizure disorder, part of his Angelman Syndrome diagnosis, would be managed in a classroom setting.

Devon had experienced several types of seizures, including myoclonic and atonic episodes, which were unpredictable and sometimes subtle. After consulting with his neurologist, the family transitioned Devon from valproic acid, which had caused tremors and regression in motor skills, to a combination of newer antiepileptic medications (i.e., levetiracetam and lamotrigine). These medications offered better seizure control and fewer side effects, allowing Devon to regain balance and coordination.

Before the school year began, Katherine and Greg requested a meeting with the preschool director, classroom teacher, and school nurse. They brought Devon's medical records, seizure action plan, and a letter from his neurologist outlining his diagnosis and treatment regimen. The family emphasized the importance of staff recognizing the signs of his seizures, which could include sudden loss of muscle tone, brief staring spells, or jerky movements.

The school nurse worked closely with the family to develop an individualized health plan (IHP) for Devon. This plan included:

- **Medication Management:** Ensuring that Devon received his antiepileptic medications at home and monitoring for any side effects during the school day.

- Seizure Response Protocols: Training staff to recognize seizure types and respond appropriately, including when to administer emergency medication and when to call 911.
- Environmental Safety: Modifying the classroom to reduce fall risks, such as using soft mats and ensuring Devon had access to supportive seating.
- Communication Strategies: Coordinating with the speech-language pathologist to ensure Devon's AAC device was accessible and that staff were trained to use it during and after seizure episodes.

The preschool staff expressed a willingness to learn and support Devon. The nurse arranged a professional development session using resources from the Angelman Syndrome Foundation to educate staff on seizure management and the unique needs of children with AS.

Throughout the school year, Katherine maintained regular communication with the nurse and teacher, sharing updates from Devon's medical team and noting any changes in his seizure activity. The collaborative approach helped the family feel empowered and reassured that Devon was safe and supported.

While the journey was not without challenges, such as adjusting to medication side effects and managing occasional nighttime seizures, the family found comfort in the school's commitment to inclusive care. Devon thrived in his classroom, forming bonds with peers and making progress in communication and motor skills, all while receiving the attentive nursing care he needed.

Discussion Prompts:

- What strategies can early childhood programs use to effectively collaborate with families and healthcare providers when managing complex medical needs like seizure disorders? How can these strategies be adapted to support children with rare genetic conditions such as Angelman Syndrome?
- What are the essential components of a seizure action plan in a preschool setting, and how can educators and support staff be trained to respond confidently and appropriately during a seizure episode?
- How can early childhood educators create a safe and inclusive environment for children with seizure disorders without compromising opportunities for exploration, peer interaction, and developmental growth?