


# Chapter 7


## Angelman Syndrome

### Neurocognitive and Linguistic Profile, Overlaps, Interventions, and Quality of Life


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#### ABSTRACT

*Angelman Syndrome (AS) is a rare neurogenetic disorder caused by a mutation or deletion of the UBE3A gene on chromosome 15, affecting the brain's development and function. Neurocognitively, individuals with AS often exhibit significant developmental delays, with limited cognitive abilities, impaired motor coordination (ataxia), and epilepsy being common. While cognitive impairment is a hallmark of the syndrome, memory and problem-solving abilities are also significantly affected. Linguistically, AS is characterized by profound speech impairments, with most individuals developing little to no functional*

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*speech. Early diagnosis, often through genetic testing, is crucial for the implementation of early intervention programs that focus on speech therapy, physical therapy, and behavioral management. Specialized interventions that address motor deficits, language development, and behavioral concerns can enhance cognitive and social functioning, although the overall prognosis remains one of lifelong disability.*

## **INTRODUCTION**

Angelman Syndrome (AS) is a rare neurogenetic disorder that primarily affects the nervous system, leading to developmental delays, motor dysfunction, and characteristic behavioral traits such as frequent laughter, hyperactivity, and a happy demeanor. First described by Dr. Harry Angelman in 1965, the syndrome was initially misdiagnosed as other forms of intellectual disability (Angelman, 1965). It is now understood that AS results from the loss of function of the *UBE3A* gene located on the maternal copy of chromosome 15, which plays a critical role in regulating protein degradation in neurons (Williams et al., 2006).

The disorder typically manifests in early childhood, with symptoms including severe developmental delays, absent speech, seizures, and ataxia. According to research by Buiting et al. (2016), AS is caused by various genetic mechanisms, such as deletions on chromosome 15q11-q13, mutations in the *UBE3A* gene, or paternal uniparental disomy. These genetic defects prevent the maternal *UBE3A* allele from being expressed in neurons, leading to the neurological symptoms observed in individuals with AS (Krzieski et al., 2024).

While there is no cure for AS, advancements in genetic research have led to potential therapeutic approaches aimed at restoring *UBE3A* function. Current interventions, such as physical, speech, and behavioral therapies, focus on improving quality of life (Tan et al., 2011). Despite these efforts, there remains much to be understood about the underlying biology of the syndrome and potential treatment strategies.

## **NEUROCOGNITIVE AND LINGUISTIC PROFILE**

Angelman Syndrome (AS) is characterized by a profound impact on neurocognitive development, with individuals experiencing severe intellectual disabilities and cognitive impairments (Megari et al., 2024). One of the hallmark features of AS is global developmental delay, with affected individuals typically achieving significant cognitive milestones much later than their peers. According to Williams et al. (2010), individuals with AS often have severe intellectual disabilities, with an average IQ below 50, though traditional IQ tests may underestimate their abilities due to significant motor and communication impairments.

Speech and language deficits are among the most distinctive neurocognitive characteristics of AS. Most individuals with AS are either non-verbal or have extremely limited speech capabilities, often limited to just a few words or none at all (Clayton-Smith, 2001). However, despite these deficits, receptive language—understanding spoken language—appears to be better preserved than expressive language, suggesting that individuals with AS may comprehend more than they can verbally express (Bonati et al., 2007).

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