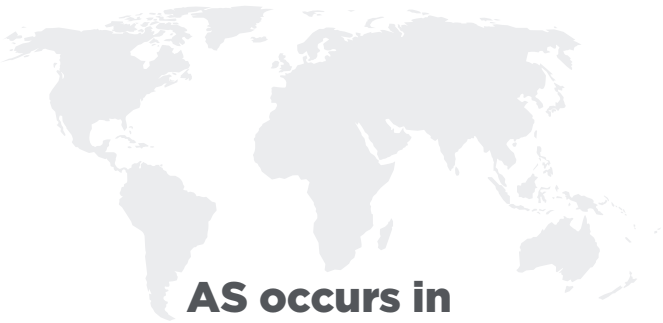
ANGELMAN SYNDROME (AS)

What is Angelman syndrome?

Angelman syndrome (AS) is a rare genetic neurological condition characterized by severe developmental delays, intellectual disability, speech impairment, problems with balance and coordination, seizures, poor sleep and behavioral issues.[1]



AS occurs in 1 in 15,000 individuals worldwide.[2]

Individuals with AS can live a normal lifespan but are unable to live independently and will require lifelong support from a caregiver.[1]

What causes AS?

AS is caused by the **loss of function** of the maternally inherited **UBE3A gene** on chromosome 15.[1]

When the UBE3A gene does not work correctly or is absent, **the UBE3A protein can not be produced,** which can cause AS.[1]

What are the signs and symptoms?

AS typically presents in infancy.

While the condition is present at birth, it is often seizures and developmental delays that are first noticed by caregivers, typically becoming apparent **before the child is 2 years old.**[1]

Signs and symptoms include:[1]

- **Developmental delays** (e.g., motor, language and cognitive functioning)
- **Difficulty with expressive and receptive communication**
- **Seizures**
- **Behavior** (including hyperactivity, irritability and anxiety)
- **Sleep challenges**
- **Gastrointestinal problems** (e.g., constipation and GERD)
- **Physical and facial features** (e.g., head size, often happy demeanor)
- **Ataxia**

How is AS diagnosed?

The **current guidelines** to diagnose AS is through:[2]

- Genetic blood testing or cheek swab
- Clinical evaluation

DNA methylation analysis (a blood test) is usually the first test done when AS is suspected.[1]

This analysis can **detect abnormalities** such as chromosomal deletion or issues with how genes are marked for expression.[1]

DNA methylation analysis detects about **80% of people** with AS.[1]

AS can be difficult to diagnose, as it shares symptoms that can be associated with other conditions like autism or cerebral palsy.[2]

Early diagnosis, intervention and treatment — including physical, behavioral and speech therapy — **can help improve quality of life for those living with AS.**[1]

What is the current standard of care for AS?

Some physical symptoms, as well as seizures, behavior and sleep, can be managed with existing medicines; however, there are **currently no approved disease modifying therapies** that address the underlying cause of the condition, help improve communication or reduce overall disease severity.[1]

Some therapies can help, including:[1]

- **Physical and occupational therapy**
  Enhances motor skills, such as walking mobility and balance
- **Speech therapy**
  Focuses on nonverbal communication through sign language and visual aids, like an Augmentative and Alternative Communication (AAC) device
- **Behavior therapy**
  Manages issues like hyperactivity and sleep disturbances. Additionally, Applied Behavior Analysis (ABA) and other behavioral strategies may be employed to improve sleep patterns, manage hyperactivity and reduce anxiety

Ongoing research into the science of AS and improvements in testing/diagnosis is being conducted to **better understand the condition** and to **develop treatments** that target the underlying cause of the disease.

For more information on AS, visit the following resources:

Angelman Syndrome Foundation

Foundation for Angelman Syndrome Therapeutics

Angelman Syndrome Alliance

References

1 Yale Medicine. Angelman syndrome. Yale School of Medicine. https://www.yalemedicine.org/conditions/angelman-syndrome. Accessed November 6, 2025.

2 Angelman Syndrome Foundation. What is angelman syndrome. https://angelman.org/about-angelman-syndrome. Accessed November 6, 2025.