

# Raising Awareness about Angelman Syndrome

Angelman Syndrome is named after Dr. Harry Angelman, an English physician who was the first to describe the characteristics of the syndrome in 1965.

## 1 Angelman Syndrome

Angelman Syndrome is a rare genetic and neurological disorder that shares similar symptoms and characteristics to those associated with autism, cerebral palsy, Prader-Willi syndrome, and other genetic disorders. Because of these similarities and the lack of general knowledge of this syndrome, it is often mistaken for one or more other disorders and misdiagnosed.

## 2 Cause

Angelman syndrome is caused by a loss of function from the UBE3A gene. People normally inherit one copy of the gene from each parent and both copies of the gene are active within the body. However, in a person with Angelman syndrome, either the maternal copy of the gene is mutated, a segment is deleted, or two copies of the paternal copy is inherited. However, there is still 10-15% of cases where the cause of the syndrome is not related to this gene and therefore unknown.

## 3 Symptoms

Symptoms of Angelman syndrome vary widely from person to person. Common symptoms include:

- developmental delays and intellectual disabilities
- no or minimal speech
- difficulty walking, moving or balancing (ataxia)
- frequent smiling /laughter and an excitable personality

Other signs associated with the syndrome are seizures, a small head size, and tongue thrusting.

## 4 Diagnosis

Since a majority of the symptoms associated with Angelman syndrome is developmental, most children are diagnosed by age 1-2. A blood test is sufficient for 80-85% of diagnoses. The remaining 15-20% should see a clinician who is familiar with the syndrome for a clinical diagnosis.

## 5 Treatment

Currently, there is no specified treatment for Angelman syndrome. A combination of physical, occupational, behavioral, and medical therapies are recommended on a case-by-case basis. Most people with Angelman syndrome have a normal life span and do not show developmental regression with age.

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## Raising Awareness

50% of individuals with Angelman's syndrome are originally misdiagnosed due to the lack of awareness surrounding the disorder. Late diagnosis can prevent individuals from early intervention and delay progress towards developmental milestones.

For more information about Angelman syndrome or to find support groups near you, visit: [www.angelman.org](http://www.angelman.org)

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## References:

<https://www.angelman.org/what-is-as/diagnosis/>

<https://www.mayoclinic.org/diseases-conditions/angelman-syndrome/symptoms-causes/syc-20355621>

<https://ghr.nlm.nih.gov/condition/angelman-syndrome#>