



## **The Arc Answers: Angelman syndrome**

### **How many people have Angelman syndrome?**

The prevalence of Angelman syndrome among adults and children is estimated to be 1 in 12,000-20,000 people. Males and females are equally likely to be affected by this neuro-genetic disorder, which is often misdiagnosed as cerebral palsy or autism.

### **What causes Angelman syndrome?**

Angelman syndrome is caused by one of several possible genetic anomalies on chromosome 15, including the deletion or mutation of genetic material. These are random events that happen during the embryonic development.

### **Prader-Willi syndrome also occurs due to anomalies on chromosome 15. What's the difference?**

Angelman syndrome is caused by either a random deletion on the chromosome 15 inherited from the mother, or the inheritance of two copies of the paternal chromosome 15 (none from his or her mother). Prader-Willi syndrome is caused when the paternal copy of chromosome 15 is either partially or entirely missing.

### **What are the primary characteristics of Angelman syndrome?**

Angelman syndrome mainly affects the nervous system. Characteristics become more noticeable between 6 months and a year of age, but most children are not diagnosed until ages 3-7. Characteristics consistently found include: developmental delay, including intellectual disabilities; movement or balance disorder, leading to uneven gait; minimal or no use of verbal language; and unique behaviors, including frequent laughter/smiling, happy demeanor, and hand-flapping or waving movements. People with Angelman syndrome use non-verbal communication methods, but are usually able to understand simple language and basic commands.

Most people with Angelman syndrome also have a smaller head circumference and seizures (average onset around age 3, severity decreases with age). Between 20-80 percent of people have additional characteristics, some of which include: a protruding tongue, feeding problems during infancy, frequent drooling, crossed eyes, lighter skin and hair color, increased sensitivity to heat, and attraction to water and shiny objects.

### **What are common medical concerns?**

People with Angelman syndrome experience relatively good physical health. However, common medical concerns include seizures of varying severity, constipation, a greater risk for scoliosis, obesity as children experience puberty and move into adulthood, and decreased mobility in later years.

### **What interventions have been proven helpful for people with Angelman syndrome?**

An individualized combination of physical, occupational, communication, and behavioral therapies have proven to help improve quality of life for people with Angelman syndrome. In addition, children and adults may benefit from the use of assistive technology that can help them communicate. As people with Angelman syndrome have an average life expectancy, services will need to be established for the long-term and re-evaluated periodically.

### **Where can I find other resources?**

Angelman Syndrome Foundation: <http://www.angelman.org/>

Resources in Spanish: <http://www.angelman.org/stay-informed/angelman-syndrome---spanish/>

Angelman, Rett, & Prader-Willi Syndromes Consortium:

<http://rarediseasesnetwork.epi.usf.edu/arpwsc/index.htm>

National Institutes of Health: <http://www.ninds.nih.gov/disorders/angelman/angelman.htm>

US National Library of Medicine: <http://ghr.nlm.nih.gov/condition=angelmansyndrome>

Madison's Foundation – Rare Disease database:

[http://www.madisonsfoundation.org/index.php/component/option.com\\_mpower/Itemid,49/diseaseID,63/](http://www.madisonsfoundation.org/index.php/component/option.com_mpower/Itemid,49/diseaseID,63/)

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