

# Angelman Syndrome

## TIPS AND RESOURCES



### **What is Angelman syndrome?**

Angelman syndrome is a genetic disorder that causes developmental delay and neurological problems. Angelman syndrome is thought to occur in about 1 in 15,000 births. The United States and Canada have an estimated 5,000-10,000 individuals living with Angelman syndrome.

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

Individuals with Angelman syndrome usually are missing a gene on the 15th chromosome called UBE3A. Sometimes UBE3A is present, but is functioning abnormally.

### **What are the effects of Angelman syndrome?**

Angelman syndrome is difficult to detect at birth. Between the ages of 6 and 12 months, developmental delays may become apparent. All individuals with Angelman syndrome have difficulty with speech and movement. Most have abnormal brain activity, seizures, and microcephaly (the circumference of the head is smaller than normal because the brain has not developed properly or has stopped growing).

Many individuals with Angelman syndrome have abnormal feeding, fair skin, a wide smile, strabismus (a condition where the eyes do not look toward the same object together), and difficulty sleeping

through the night. Many walk with a wide puppet-like gait and their arms outstretched. Recent research shows that about half of individuals with Angelman syndrome also will show signs of autism spectrum disorder.

Outbursts of laughter are frequent among individuals with Angelman syndrome. Happiness seems to be a constant state, and social smiling may be prevalent. Many individuals are very social and have excellent memories for faces and places.

### **Can Angelman syndrome be treated?**

Early diagnosis and early intervention is the best treatment. Most individuals with Angelman syndrome make steady developmental progress and do not regress. Those who experience seizures usually need medical care. Physical, occupational, speech, and behavioral therapies contribute to improving the quality of life. Water play seems to be especially appealing to most individuals with Angelman syndrome, so swim therapy is often a favorite option. Another option may be hippotherapy, a therapeutic approach that uses horses instead of typical physical therapy equipment.

It is unlikely that individuals with Angelman syndrome will live independently, but encouraging independence as much as possible is beneficial. Individuals with Angelman syndrome learn best through repetition and structure. Plan well and make learning a game.

### **What are the special needs of families?**

It may be beneficial for a family with a member with Angelman syndrome to seek group support. Membership in the Angelman Syndrome Foundation and in the Foundation for Angelman Syndrome Therapeutics helps families get information on the latest treatments and therapies, and also helps them to connect with others in similar situations. Many issues experienced by people with Angelman syndrome are similar to what people with autism spectrum disorders face, so membership in a local chapter of The Arc or Autism Society can be helpful for learning about special education law, local programs, and new therapies.

*Please see reverse for information about Vanderbilt Kennedy Center and other programs.*

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## How We Can Help

### Who We Are and Who We Serve

The **Vanderbilt Kennedy Center (VKC)** works with and for people with disabilities and family members, service providers and advocates, researchers and policy makers. It is among only a few centers nationwide to be both a **University Center for Excellence in Developmental Disabilities Education, Research and Service** and a national **Eunice Kennedy Shriver Intellectual and Developmental Disabilities Research Center**. The VKC also is a member of the **Rare Diseases Clinical Research Network**, a national network that facilitates collaboration among experts in many different types of rare diseases.

### Tennessee Disability Pathfinder

Pathfinder provides a telephone helpline, web, and print resources in English and Spanish that connect the Tennessee disability community with service providers and resources. Pathfinder is a project of the VKC and the Tennessee Council on Developmental Disabilities. See [www.familypathfinder.org](http://www.familypathfinder.org), (615) 322-8529, toll-free (800) 640-4636.

### Vanderbilt Kennedy Family Outreach Center

This center provides families with a single point of entry into the many services and supports of the VKC, Vanderbilt University, and the community. Services include research projects, clinics, social work services, and recreational and arts programs. Contact [familyoutreach@vanderbilt.edu](mailto:familyoutreach@vanderbilt.edu), (615) 936-5118.

### Two Ways to Take Part in Research

The VKC serves families through research studies. **StudyFinder**, a searchable database, lists studies seeking people of all ages with and without disabilities. See [kc.vanderbilt.edu/studyfinder](http://kc.vanderbilt.edu/studyfinder). **Research Family Partners** allows families with and without disabilities to register and to be contacted when a study begins for which a family might be eligible. Contact [kc.vanderbilt.edu/RFP](http://kc.vanderbilt.edu/RFP), (615) 936-0448, toll-free (866) 936-8852.

### Alphabet Therapy Research

VKC researchers are focusing on a new way to help school-aged children with Angelman syndrome learn the alphabet. The hope is that by learning the alphabet, these children will be able to communicate better. They are using a technique called Alphabet Therapy, which is a combination of Applied Behavior Analysis ideas and a quick prompting method. For information about Alphabet Therapy, contact Terry Jo Bichell at [terry.jo.bichell@vanderbilt.edu](mailto:terry.jo.bichell@vanderbilt.edu), (615) 322-8093.

### SibSaturdays

This program provides siblings who have a brother or sister with a disability with the opportunity to play games, meet friends, and participate in group discussions. Contact [familyoutreach@vanderbilt.edu](mailto:familyoutreach@vanderbilt.edu), (615) 936-5188.

### Behavior Analysis Clinic

This clinic provides comprehensive health, educational, and behavioral assessments to identify causes of behavioral problems and to develop person-centered interventions. The clinic serves children ages 5-18. Contact [n.houchins-juarez@vanderbilt.edu](mailto:n.houchins-juarez@vanderbilt.edu), (615) 322-9007.

### Other Local and National Resources

**Monroe Carell Jr. Children's Hospital at Vanderbilt Health Library, Jr. League Family Resource Center**, [www.vanderbiltchildrens.com/healthlibrary](http://www.vanderbiltchildrens.com/healthlibrary) (615) 936-2558, toll-free (800) 288-0391

#### Genetics Clinic

[www.vanderbiltchildrens.com/interior.php?mid=178](http://www.vanderbiltchildrens.com/interior.php?mid=178) (615) 322-7601

#### Pediatric Neurology Clinics

[www.vanderbiltchildrens.com/interior.php?mid=595](http://www.vanderbiltchildrens.com/interior.php?mid=595) (615) 936-5536

### Angelman Syndrome Foundation

[www.angelman.org/angel](http://www.angelman.org/angel), (800) 432-6435

### Foundation for Angelman Syndrome Therapeutics

[www.cureangelman.org](http://www.cureangelman.org), (866) 783-0078

### Autism Society of America, [www.autism-society.org](http://www.autism-society.org)

**Autism Society of Middle Tennessee**, [www.tnautism.org](http://www.tnautism.org)

### Saddle Up!

[www.saddleupnashville.org](http://www.saddleupnashville.org), (615) 794-1150

### Tennessee's Early Intervention System (800) 852-7157

### Tennessee Developmental Disabilities Network

[www.tennddnetwork.org](http://www.tennddnetwork.org)

### The Arc US, [www.thearc.org](http://www.thearc.org), (301) 565-3842

**The Arc of Tennessee**, [www.thearc.org](http://www.thearc.org), (800) 835-7077 (615) 248-5878. See also Tennessee chapters

### Rare Diseases Clinical Research Network

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

### National Institute of Neurological Disorders and Stroke

[www.ninds.nih.gov/disorders/angelman/angelman.htm](http://www.ninds.nih.gov/disorders/angelman/angelman.htm)

### Contact the Vanderbilt Kennedy Center

Nashville (615) 322-8240

Toll-Free (866) 936-VUKC [8852]

[www.kc.vanderbilt.edu](http://www.kc.vanderbilt.edu)

[kc@vanderbilt.edu](mailto:kc@vanderbilt.edu)



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FOR EXCELLENCE IN DEVELOPMENTAL DISABILITIES