Angelman Syndrome

TIPS AND RESOURCES FOR FAMILIES

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 resources.

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RESOURCES

Who We Are and Who We Serve

The **Vanderbilt Kennedy Center** (VKC) works with and for people with disabilities and their family members, educators and service providers, researchers, students, and policy makers. Faculty and staff engage in interdisciplinary research, training, service, and information dissemination and work in collaboration with local, state and national networks and partners. (615) 322-8240, toll-free (866) 936-8852, vkc.vumc.org

Tennessee Disability Pathfinder

Provides free information, referral sources, and help with navigating services via phone, email, and website. Assistance is available to individuals of all ages, all types of disabilities, and all languages spoken. Its website database has more than 3,000 agencies searchable by Tennessee county and service. Pathfinder is a project of the VKC and is partially funded by Tennessee Council on Developmental Disabilities and other state agencies. (615) 322-8529, toll-free (800) 640-4636, **DisabilityPathfinder.org**

Two Easy Ways to Take Part in Research

StudyFinder is a searchable database that lists current VKC studies. Studies seek people of all ages with and without developmental disabilities. See **vkc.vumc.org/studyfinder**. Research Match is a secure place for volunteers and researchers to connect. See **researchmatch.org**

Sibling Supports

Support for siblings who have a brother or sister with a disability, chronic health care issue, or mental health concern. SibSaturdays, ages 5 to 12 and Tennessee Adult Brothers and Sisters (TABS), ages 18+. See **vkc.vumc.org/sib**

Local and National Resources

- Angelman Syndrome Clinic
  www.childrenshospitalvanderbilt.org/clinic/angelman-syndrome
- Angelman Syndrome Foundation
  www.angelman.org
- Autism Society of America
  www.autism-society.org
- Autism Tennessee
  www.autismtn.org
- Foundation for Angelman Syndrome Therapeutics
  www.cureangelman.org
- Monroe Carell Jr. Children's Hospital at Vanderbilt Health Library, Jr. League Family Resource Center
  www.childrenshospitalvanderbilt.org/information/junior-league-family-resource-center
- National Institute of Neurological Disorders and Stroke
  www.ninds.nih.gov/Disorders/All-Disorders/Angelman-Syndrome-Information-Page
- Pediatric Genetics and Genomic Medicine
  www.childrenshospitalvanderbilt.org/service-line/pediatric-genetics-and-genomic-medicine
- Rare Diseases Clinical Research Network
  www.rarediseasesnetwork.org/diseases#FAD2
- Saddle Up!
  www.saddleupnashville.org
- Tennessee's Early Intervention System
  www.tn.gov/didd/for-consumers/tennessee-early-intervention-system-teis.html
- The Arc of Tennessee
  www.thearctn.org

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