About Angelman Syndrome

Angelman Syndrome (often abbreviated AS) is a severe neurological disorder affecting approximately 1 in 15,000 live births. AS is characterized by profound developmental delays, problems with motor coordination (ataxia) and balance, and epilepsy. Individuals with AS do not develop functional speech; however, some parents have reported limited word approximations. The seizure disorder in individuals with AS can be difficult to treat. Feeding disorders in infancy are common and some persist throughout childhood. Sleeping difficulties are commonly noted in individuals with AS. AS affects all races and both genders equally.

Individuals with AS tend to have a happy demeanor, characterized by frequent laughing, smiling and excitability. Many individuals with AS are attracted to water and take great pleasure in activities like swimming and bathing.

AS may be misdiagnosed as cerebral palsy or an autistic spectrum disorder. If an individual fits the diagnostic criteria, testing for AS is recommended as medical care and educational strategies that benefit individuals with AS can be drastically different than those recommended for other disorders in spite of seemingly similar characteristics. For information on AS testing, please visit www.CureAngelman.org/what.html.

If AS is not confirmed through genetic testing, there are other syndromes that share characteristics with AS and should be considered in making a correct diagnosis. For a list of mimicking disorders, please visit www.CureAngelman.org/what.html

The cause of AS is known. AS is a single gene disorder that results in the loss of functional levels of maternal UBE3A protein in the brain. The paternal UBE3A gene is present in AS, but is not expressed due to a genetic mechanism called imprinting. There are preeminent animal models of AS available for testing and experiments. AS has been genetically cured in the animal model. Paternal UBE3A in mouse has been activated in the laboratory. Adding UBE3A into the brain in the adult mouse model reversed the symptoms of the disorder. Thus, the groundwork for uncovering viable treatments for AS is well established. FAST is focused on bringing practical treatments into current medical practice as efficiently as possible by supporting a robust research agenda, and by promoting unprecedented cooperation among leading AS experts.

Diagnostic Criteria for Angelman Syndrome

Findings typically present in affected individuals

- Normal prenatal and birth history, normal head circumference at birth, no major birth defects.
- Normal metabolic, hematologic, and chemical laboratory profiles.
- Structurally normal brain by magnetic resonance imaging (MRI) or computed tomography, although mild cortical atrophy or dysmyelination may be observed.
- Delayed attainment of developmental milestones without loss of skills.
- Evidence of developmental delay by age 6-12mths, eventually classified as severe.
- Speech impairment, with minimal to no use of words; receptive language skills and nonverbal communication skills higher than expressive language skills.
- Movement or balance disorder, usually ataxia of gait and/or tremulous movement of the limbs.
- Behavioural uniqueness, including any combination of frequent laughter/smiling; apparent happy demeanor; excitability, often with hand-flapping movements; hypermotoric behavior; short attention span.

Findings in more than 80% of affected individuals

- Delayed or disproportionately slow growth in head circumference, usually resulting in absolute or relative microcephaly by age 2 yrs.
- Seizures, usually starting before age 3 years.
- Abnormal electroencephalogram (EEG), with a characteristic pattern of large-amplitude slow-spike waves.

Findings in fewer than 80% of affected individuals

- Flat occiput.
- Occipital groove.
- Protruding tongue.
- Tongue thrusting; suck/swallowing disorders.
- Feeding problems and/or muscle hypotonia during infancy.
- Prognathia.
- Wide mouth, wide-spaced teeth.
- Frequent drooling.
- Excessive chewing/mouthing behaviours.
- Strabismus.
- Hypopigmented skin, light hair and eye color (compared with family); seen only in those with a deletion.
- Hyperactive lower-extremity deep-tendon reflexes.
- Uplifted, flexed arm position especially during ambulation.
- Wide-based gait with out-going (i.e., pronated or valguspositioned) ankles.
- Increased sensitivity to heat.
- Abnormal sleep-wake cycles and diminished need for sleep.
- Attraction to/fascination with water; fascination with crinkly items such as certain papers and plastics.
- Abnormal food-related behaviors.
- Obesity (in the older child; more common in those who do not have a deletion).
- Scoliosis.
- Constipation.

From: "Genetics in Medicine:"Clinical and genetic aspects of Angelman Syndrome." Charles A Williams, MD, Daniel J Driscoll, PhD, MD and Aditi I. Dagli MD. May 2010
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CUREAngelman.org

FAST
Foundation for Angelman Syndrome Therapeutics

Angelman Syndrome (AS) is a neurodevelopmental disorder affecting approximately 1 in 15,000 live births.

Although the cause of AS is known, there are currently no treatments available for this disorder. FAST is committed to funding the research that will lead to treatments and eventually a cure.

Learn about AS and why we believe a cure is within reach...

CUREAngelman.org

Foundation for Angelman Syndrome Therapeutics

Raising Hope Through Research

Changing Lives Together

About FAST

The Foundation for Angelman Syndrome Therapeutics ("FAST") is an organization of families and professionals dedicated to finding a cure for Angelman Syndrome and related disorders through the funding of an aggressive research agenda, education, and advocacy.

FAST is committed to funding cutting-edge research in order to discover therapeutic strategies to cure Angelman Syndrome. FAST is served by two boards: the Board of Directors and the Scientific Advisory Board. Together, we strive to bring practical treatment into current medical practice as quickly as possible.

Board Of Directors

Our all-volunteer Board of Directors, almost all of whom are parents of a child with AS, utilize their expertise in various fields to advance our mission, assess our strategies and create opportunities that aggressively pursue our goals.

Scientific Advisory Board

The Scientific Advisory Board consists of nationally and internationally recognized clinicians and scientists considered experts in their respective fields. Scientific Advisory Board members review all applications for funding and their critiques and recommendations are brought to the board by the Chief Science Officer. The depth and breadth of knowledge encompassed by our Scientific Advisory Board members allows for expert evaluation of proposals from advancing basic science to improving clinical practice.

To learn more about FAST, our mission, and the professionals working towards our goal, please visit www.CureAngelman.org/about.html

Research

FAST-funded research has already identified one potential treatment for several of the symptoms of AS. In near unprecedented fashion, FAST then funded a human clinical trial for this compound, taking a potential treatment from the laboratory to patients in just one year.

To continue our search for therapeutics, the FAST Integrative Research Environment (FIRE) was established. FIRE is a coordinated consortium of leading researchers working together to identify, characterize and implement new therapeutics for the treatment and ultimate cure of AS. By bringing together prominent AS researchers to not only collaborate but also exchange information in real-time, FIRE will exponentially escalate the journey towards a cure.

In addition to FIRE, our grants program enables individual researchers to pursue projects targeted specifically towards our goal of finding a treatment for Angelman Syndrome. Our expedited review process ensures funds are put into quality research as efficiently as possible. FAST-TRAC Grants and Grants-in-Aid are reviewed on a rolling basis with no application deadline.

Through our fellowship program, FAST has funded over $360,000 in postdoctoral fellowships to support exceptional early career scientists working on Angelman Syndrome. By supporting postdoctoral fellows, we are increasing the number of outstanding researchers focusing on AS.

To learn about the research funded by FAST and FAST's funding philosophy, please visit www.CureAngelman.org/research-funded.html

GET INVOLVED

FAST is focused on one goal: developing treatments for, and ultimately curing, Angelman Syndrome. Your involvement is critical in helping us reach this goal as quickly as possible.

Membership in FAST is free and ensures that you stay current on all the latest research and progress. You will receive email updates as well as our e-newsletter "Get Informed FAST" to keep you up-to-date on all fronts.

Be sure to "Like" us on Facebook at www.facebook.com/www.cureangelman.org
Follow us on Twitter at twitter.com/CureAngelman and on Pinterest at pinterest.com/cureangelman/
Read about us and our community on blog.cureangelman.org

Ways to Give

Make a one-time donation or a recurring gift, give a charitable donation in lieu of a gift, as an Honor or Memory Gift, or leave a lasting legacy through support of an endowment fund.

One of the smartest ways to amplify your donation to FAST is through a Corporate Matching Gift Program. Check with your company's Human Resources office to see if they offer gift matching. If they don't, ask them to start.

To make a donation to FAST, visit www.CureAngelman.org/donate.aspx or scan the QR Code at the right.

Ways to Fundraise

No matter what you enjoy doing, from bake sales to Zumbathons , website search engines to Marathons, you can do it for FAST and the Angelman Cause.

To learn more about fundraising for FAST, please visit www.CureAngelman.org/involved.html or send your idea in an email to events@CureAngelman.org
Join us and be part of the miracle!