YOU ARE NOT ALONE

You know they hear you, but they can't form the words to answer. Walking is halted and awkward, but their energy propels them forward. And their smile seems ever-present. This is a brief portrait of Angelman syndrome, and if your family is affected by it, you need not face this journey alone.

Families and professionals have joined together to give strength and a voice to your needs and concerns. The Angelman Syndrome Foundation is dedicated to providing information, educational opportunities and support to those who need our help. The Foundation is made up of regional and local groups that may be close to you, and we strive to help you cope with and understand this special person who has touched your life. Together, we give your child a reason to smile.

THE FOUNDATION FOR KNOWLEDGE AND CONNECTION

Why does the syndrome occur? How many individuals are affected? What do we really know, and how can we learn more? Where do you turn for help and support?

The Angelman Syndrome Foundation was established in 1990 to help answer these questions and provide a network of information and support resources across the country. This pamphlet presents basic information that may help you begin your journey to understanding.

ANGELMAN SYNDROME WEBSITES

Angelman Syndrome Foundation (USA)
www.angelman.org

Casa Angelman (Argentina)
www.casaangelman.org

National Association (Argentina)
www.angelman-asa.org

Angelman Syndrome Association of Australia
www.angelmansyndrome.org

Angelman Verein Osterreich (Austria)
www.angelman.at

Comunidade Sindrome de Angelman (Brazil)
www.acsa.org.br

CASS - Canadian Angelman Syndrome Society
www.angelmancanada.org

Fondation du Syndrome Angelman du Québec
www.angelman.ca

Angelman CZ Občanské sdružení (Czech Republic)
www.angelman.cz

Angelman Foreningen (Denmark)
www.angelmanforening.dk

Angelman (Finland)
www.angelman.fi

Association Française du Syndrome d'Angelman (AFSA) (France)
www.angelman-afsa.org

Syndrome Angelman France
www.syndromeangelman-france.org

Angelman e.V (Germany)
www.angelman.de

Angelman Ireland
www.angelman.ie

Israeli Association Angelman syndrome
www.angelman.co.il

Angelman Onlus (Italy)
www.associazioneangelman.it

Organizzazione Sindrome di Angelman (Italy)
www.sindromediangelman.org

Japan
www5f.biglobe.ne.jp/~angel-no-kai

Nina Foundation (Netherlands)
www.ninafoundation.eu

Angelman Syndroom (Netherlands)
www.angelmansyndroom.nl

The Angelman Network (New Zealand)
www.angelmannetwork.com

Norsk Forening for Angelmans Syndrom (Norway)
www.angelman.no

Associação de Síndrome de Angelman de Portugal
www.angel.pt

South Africa
http://angelkids.webs.com

Asociación Síndrome de Angelman (Spain)
www.angelman-asa.org

Angelman Verein Schweiz (Switzerland)
www.angelman.ch

Angelman Syndrome Support Education & Research Trust (ASSERT) (United Kingdom)
www.angelmanuk.org

PLEASE CONTACT ME ABOUT THE FOLLOWING:

(Please check all that apply):

- ❒ Angelman syndrome
- ❒ The Angelman Syndrome Foundation
- ❒ Meetings and events
- ❒ Angelman syndrome research
- ❒ Donating to the Angelman Syndrome Foundation

Name: _____

Address: _____

City: _____

State/Province: _____

Zip/Postal Code: _____

Country: _____

Phone: _____

Fax: _____

E-mail: _____

Mail to: Angelman Syndrome Foundation
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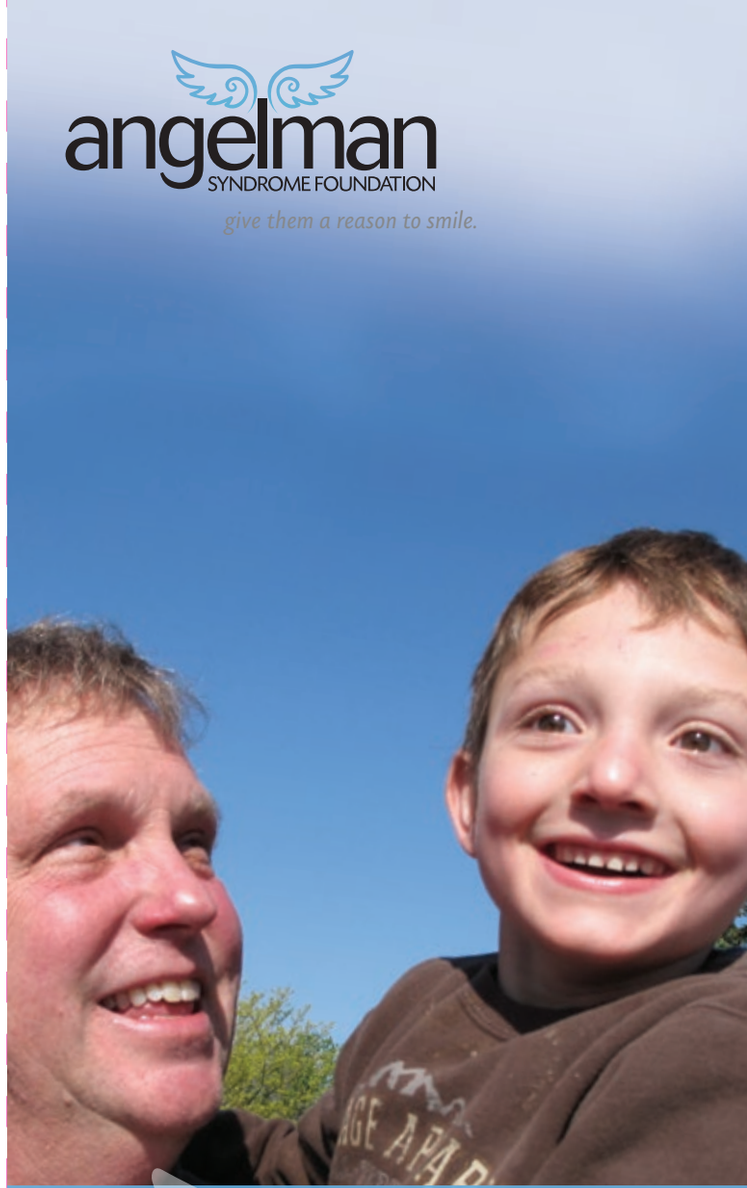
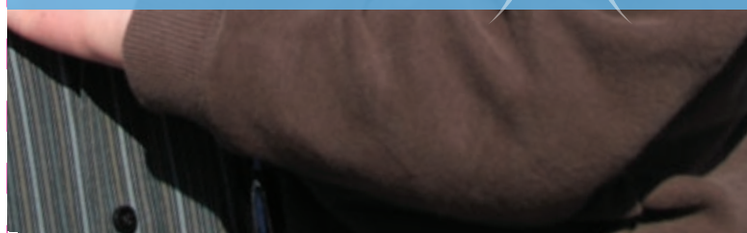


THE FACTS
ABOUT ANGELMAN SYNDROME



WHAT DO WE KNOW?

Angelman syndrome has confounded both the medical community and parents of individuals with Angelman syndrome for hundreds of years. Initially presumed to be rare, it is now believed that thousands of cases have gone undiagnosed or misdiagnosed as cerebral palsy, autism, or other childhood disorders.

HOPE THROUGH DIAGNOSIS AND RESEARCH

The first glimmer of hope for diagnosis and ultimately care and treatment of Angelman syndrome came as a result of the work of Dr. Harry Angelman in 1965. The following are clinical characteristics of Angelman syndrome:

A. Consistent (100% of cases)

- Developmental delay - functionally severe.
- Speech impairment - no or minimal use of words, receptive and non-verbal communication skills higher than verbal ones.
- Movement or balance disorder - usually ataxia of gait and/or tremulous movement of limbs. Movement disorder can be mild. May not appear as frank ataxia but can be forward lurching; unsteadiness; clumsiness; or quick, jerky motions.
- Behavioral uniqueness - any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with hand flapping movements; hypermotoric behavior.

B. Frequent (more than 80% of cases)

- Delayed, disproportionate growth in head circumference - usually resulting in microcephaly (absolute or relative) by age two. Microcephaly is more pronounced in those with 15q11.2q-13 deletions.
- Seizures - onset usually at less than three years of age. Seizure severity usually decreases with age, but the seizure disorder lasts throughout adulthood.
- Abnormal EEG - characteristic pattern with large-amplitude slow-spike waves. The EEG abnormalities can occur in the first two years of life, can precede clinical features, and are often not correlated to clinical seizure events.

C. Associated (20-80% of cases)

- Strabismus
- Hypopigmented skin - light hair and eye color when compared to family; seen only in deletion cases
- Tongue thrusting, suck/swallow disorders
- Hyperactive lower-extremity deep tendon reflexes
- Feeding problems and/or truncal hypotonia during infancy
- Uplifted, flexed arms, especially during walking
- Wide-based gait
- Prominent mandible
- Increased sensitivity to heat
- Wide mouth, widely spaced teeth
- Abnormal sleep-wake cycles and diminished need for sleep
- Frequent drooling, protruding tongue
- Attraction to or fascination with water; fascination with crinkly items such as papers and plastics
- Excessive chewing or mouthing behaviors
- Flat back of head
- Occipital groove
- Abnormal food-related behaviors
- Obesity (in the older child)
- Scoliosis
- Constipation

Today, we know that Angelman syndrome is a genetic disorder caused by abnormal function of the gene UBE3A, located within a small region (q11-q13) on chromosome #15. This region is deleted from the maternally derived chromosome #15 in approximately 70% of individuals with Angelman syndrome. For the remaining 30%, genetic testing can often identify other abnormalities that disrupt UBE3A function. Some individuals in this latter group, however, still have apparently normal genetic laboratory studies. For these individuals, the diagnosis is based solely upon clinical findings.

WHAT CAN YOU DO?

Early diagnosis and intervention are beneficial when Angelman syndrome is suspected. Diagnosis can now be accomplished within the first year after birth. Therefore, a physician familiar with Angelman syndrome can be an important resource. The Angelman Syndrome Foundation can provide assistance in connecting families and professionals interested in Angelman syndrome.

As Angelman syndrome research has accelerated, educational and behavioral interventions have been shown to be effective in the areas of communication, education, sleep disturbances and general behavior. Physical and occupational therapies, speech and language interventions, behavior modification, and parent training have proven effective. Alternative/enhanced communication techniques are very effective in individuals with Angelman syndrome, as these individuals demonstrate a much greater receptive-language ability than expressive ability.

Angelman syndrome affects males, females and all racial/ethnic groups equally. The prevalence among children and young adults is 1 in 15,000, and there are estimated to be as many as 5,000 cases in the U.S. and Canada.

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give them a reason to smile.

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Facebook.com/AngelmanSyndromeFoundation

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