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Dear Readers of *Angelman Syndrome A to Z*,

I want to sincerely thank all the family members and professionals who took time to share their invaluable information, tips, and anecdotes for the 2020 edition of *Angelman syndrome A to Z*.

As Audrey Angelman once said,

“The breaking of that awful feeling of isolation is the greatest gift one Angelman family can give to another.... Those of you who will share your experiences will do it because in the Angelman family, that is what we do.”

Even as an OLD parent of a thirty-eight year-old daughter with Angelman syndrome, it was an exhilarating experience for me to read all of your heartfelt contributions. My family joined the “pioneers” of Angelman syndrome in 1992, and never in our wildest dreams could we have foreseen the astounding strides the ASF has made since then. From funding ten million dollars in phenomenal research projects, to providing a wealth of family support services, to opening AS Clinics across the country, to hosting fifteen family conferences and numerous scientific symposiums, to coordinating annual Walks at nearly fifty sites... The ASF has accomplished all of this and so much more!

As has always been the case, the goal of the 2020 edition of *Angelman Syndrome A to Z* was to create an all-inclusive document on Angelman syndrome. Naturally, that is a HERCULEAN task! Our latest efforts are only “a tip of the iceberg”! However, I hope you will find *Angelman syndrome A to Z* to be an invaluable tool. The project’s primary design is to help you find what you are looking for quickly and efficiently.

***Professional information is listed first after each topic. Following that is information provided by the “**real experts**”... *the Angelman family members!* Some contributors chose to be anonymous, while others provided additional information in an effort to provide a reference point.

I hope you will benefit, as much as I have, from the wisdom of others in our Angelman syndrome community.

My love to you all,
Alice Evans

DISCLAIMER

Please keep in mind that many of the ideas/tips contributed by family members in *Angelman Syndrome A to Z* are merely suggestions and may not be effective for your angel or family. This reference material is NOT meant to be a medical guide or detailed scientific journal, and is not intended to replace medical treatment and/or consultation. Please verify all medical information with your health care professional. Additionally, the Angelman Syndrome Foundation does not endorse any agencies, products or services listed in this book.

A

AAC AND TECHNOLOGY

See I-iPads

Websites:

PraacticalAAC.org

Carole Zangari, PHD, CCC/SLP

AACIntervention.com and aacgirls.blogspot.com

By Caroline Musselwhite, PHD, CCC/SLP

Assistive Technology:

Proloquo2Go

<https://www.assistiveware.com/products/proloquo2go>

Tobii Dynavox

Provider of speech generating devices and symbol-adapted special education software used to assist individuals in overcoming their speech, language and learning challenges. Solutions designed to help individuals who have complex communication and learning needs participate in the home, classroom and community. The Tobii product line includes Tobii Gaze Viewer, which allows you to record real eye tracking data from any application, whether from the Internet or e-books, games, movies and more.

Mayer-Johnson

Mayer-Johnson Boardmaker Software Family is best known for creating printed and on-screen materials, like communication boards, symbol-adapted books, flashcards, schedules and more. Mayer-Johnson boasts thousands of unique special education tools that meet a wide range of goals.

Nova Chat by Saltillo

These portable communication systems with Chat software are based on an Android platform. Nova Chat devices have a variety of vocabulary configurations, thousands of symbols, voice options are portable and lightweight. Saltillo Corporation's mission is to allow people with disabilities to participate in and enjoy life to the fullest.

Aut 2 Communicate

The goal of Aut 2 Communicate is to contribute towards empowering individuals who have expressive language difficulties with a means to communicate and interact with their friends, loved ones and community; to enable them to have input into the decisions that shape their lives; to give them the voice they deserve.

Axistive

Axistive is the world's leading news portal for assistive technology, providing a wide range of free services including product reviews, industry news, market trends, workshops, downloads, and product and organization information.



Enabling Devices

Enabling Devices is a company dedicated to developing affordable learning and assistive devices to help people of all ages with disabling conditions.

Spectronics Blog

This blog lists and updates AAC Apps for iPhone/iPod touch/iPad.

The Parent Educational Advocacy Training Center (PEATC) IEP Checklist iPhone App

The IEP is an Individualized Education Program designed to support the educational needs of school aged students with disabilities. The IEP Checklist App helps parents of students with special needs become better-informed advocates by making IEP information easier to access.

Family Center on Technology and Disability

The Center on Technology and Disability (FCTD) is a resource designed to support organizations and programs that work with families of children and youth with disabilities. FCTD offers a range of information and services on the subject of assistive and instructional technologies.

Computer & Software

BigKeys

Greystone Digital, Inc. provides BigKeys keyboards which are standard sized with very large keys.

The Learning Company

The Learning Company develops and retails a full-range of fun and educational software, workbooks, flashcards and multimedia systems.

Angelman Family Contributions: AAC and Technology

We went all in on the PODD system back in 2015. I went to PODD training bought the PODD system discs, Boardmaker discs, (waterproof pages, bookbinding machine, book covers, spiral connectors etc.) and made several books. I also used 3-ring binders and made a couple books that had a built-in stands.

AS Family Member

I really tried with PODD. I found it very challenging. I found my daughter did not have good fine motor control or the ability to isolate her pointer finger, so she would rake her hand across the page. In training they tell you to presume competence and to respond to what they point to, but I couldn't usually tell what she was trying to point to, so it was very frustrating for her and me.

AS Family Member

She seemed to like videos on YouTube for kids. Roller coasters, kids making things, music videos, Kidz Bop, Masha and the Bear, Baby Shark, etc.

AS Family Member

I spoke with her school SLP and we thought trying GoTalk with her might be more motivating, as you press the button and get a verbal response. Also, the sections are in frames, so it isolates each picture. She responded better to the higher tech, interactive system; however, the record button on the back proved to be too much of a distraction, so she would end up erasing the recordings. You also have to push down pretty firmly on each box, and she did not have the fine motor control to isolate and push with purpose.

AS Family Member

When PODD was available as an app on the iPad, I bought it. I bought two iPad pros, one for her to use and one for me to model with her. I sent it to school with her, her SpEd teacher went to PODD training and was having success with her, but her teacher was the only person that understood it and she couldn't be with Caroline all the time at school. The school used Proloquo2go and there was resistance to learning a different way. Since I had a very hard time modeling the program with her and found the way it was organized confusing, I didn't push to have them learn PODD.

AS Family Member

A friend, Sandra, had LAMP on an iPad for her Angel. We tried it, under her account, but that account was set up with too many small icons. You could reduce the amount of icons that you could see on the page by hiding them, but because it was set up for her daughter, we couldn't change the amount of the boxes on the page. My daughter has vision issues, so this did not work for us.

AS Family Member

Last Christmas (2018) we started Caroline on the Proloquo2go system that the school uses. We have had much more success with this system. I understand and can navigate it very easily, so I use it all the time at home which reinforces to Caroline that this is her voice.

AS Family Member

Bottom line, try many different options, see what works best for your child and your family. We have the iPads for her to talk, but we have laminated pages with Velcro pictures on them in the shower, for the pool, for outside water play, etc. Also, YOU have to learn and use the system. It won't happen if only the teachers or therapists are working with your child on the program. YOU must reinforce the learning at home. It takes time.... but it is so worth it to give your Angel the ability to make their own choices and to be able to communicate their needs to a stranger in the event of an emergency.

Donna, sdangelmom@gmail.com, angel Caroline Del+

Our son has done very well learning the LAMP AAC program for the iPad. It is a motor-planning concept that works well for kids with Autism. He has learned where words are located and only has to push 1-2 icons to make a word appear in the text line and hear it voiced. We have enjoyed knowing what he's interested in and being able to verbally interact, and he often likes to make funny comments!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18 Del+ Class 1

ACID REFLUX (Also See G-GERD)

Some parents of angels use medications to treat GERD (gastroesophageal reflux disease). A few examples are: Nexium, Protonix, Prilosec, and Prevacid. *** Always get advice from your doctor!

Angelman Family Contributions: Acid Reflux

Avoiding dairy is one of the best treatments for my son's acid reflux.

AS Family Member

Our son's acid reflux became worse as he got older. He finally developed a gastro bleed incident and was started on Nexium, which he takes now to avoid a recurrence. He gags easily and tends to vomit when he has nasal discharge into his throat.

Susan Pike

ADULT GUARDIANSHIP

Watch the ASF Educational Webinar on Guardianship, presented by Dr. Eric Wright.
<https://www.youtube.com/watch?v=qhdj-2jRaSc&feature=youtu.be>

See [National Guardianship Association](#) Affiliates by State.

Angelman Family Contribution: Guardianship

On my angel's 18th birthday we went to court to become our daughter's legal guardians. This is a legal necessity with HIPAA and dealing with governmental agencies on our young adult's behalf. Make SURE to check all designated beneficiaries for all insurance policies etc... so as not to negate any governmental benefits from an inheritance, etc... This is also where a special needs trust is very beneficial for our angel's future wellbeing.

Kathy, Fort Wayne, Indiana, angel Juliana (UPD)

ADULT HEALTH ISSUES

[Click here to access the American Journal of Medical Genetics paper.](#)

Anna M. Larson Julianna E. Shinnick Elias A. Shaaya Elizabeth A. Thiele Ronald L. Thibert

First published: 26 November 2014 <https://doi.org/10.1002/ajmg.a.36864> Cited by: 21

Dr. Ron Thibert, the well-known Angelman syndrome clinician and champion of the low-glycemic index dietary seizure treatment, and Dr. Anna Larson, both of Massachusetts General Hospital, published the findings from their clinical investigation into health issues that adults with AS experience.

The research team conducted standardized phone interviews with caregivers for 110 adolescents and adults with AS aged 16 to 50 years old. The impact of age, gender, and genotype on specific outcomes in neurology, orthopedics, internal medicine, and psychiatry were investigated, but did not address treatment. Further work should continue to refine the observable characteristics of older individuals with AS. Primary areas of clinical management identified in this research include seizures, sleep, aspiration risk, GERD, constipation, dental care, vision, obesity, scoliosis, bone density, mobility, communication, behavior, and anxiety.

The following is a summary of their findings.

Active Seizures

- Present in 41% of individuals
- Epilepsy severity may assume a bimodal age distribution: seizures are typically most severe in early childhood but may recur in adulthood

Sleep Dysfunction

- Present in 72% of individuals
- Late-adolescent and adult sleep patterns are improved when compared to the degree of sleep dysfunction present during infancy and childhood
- However, prevalence of poor sleep in adults remains quite high

Significant Constipation

- Present in 85% of individuals

Overweight / Obesity

- Present in 32% of individuals, with obesity disproportionately affecting women

Scoliosis

- Affects 50% of individuals with an average age of diagnosis at 12 years old
- 24% of those diagnosed with scoliosis required surgery, an intervention disproportionately affecting men

Walking

- 68% are able to walk independently

Speaking

- 13% are able to speak 5 or more words

Self-Injurious Behavior

- 52% of individuals exhibit self-injurious behavior

ADULT PROGRAMS

Angelman Family Contributions: Adult Programs

When selecting an adult day program for your angel, don't be afraid to look outside your community. Tour as many programs as you can before making a selection. Know that your decision does not have to be final either. If at any point, you don't feel comfortable, you have the right to change programs. Also, make friends with the staff members at the program. They will be your best allies into your Angel's day.
Sandra, sandra1rivero@gmail.com, angel Amanda, age 23, UPD

Our daughter attends a day program for adults. We quickly learned at the beginning of the process that NO program we visited met our very high expectations. However, we chose the best one we could find and came to realize that the benefits far outweighed any reservations we had. The key is to ensure that your child has an exceptional teacher or aide and communicates daily in a "journal". I also write daily uplifting notes and convey our gratitude. These underpaid professionals work extremely hard!

Alice, sandiegoasfwalk@gmail.com, Whitney, age 38, Deletion+ Class 1

ADVOCACY

ASF Family Resource Team Member

[Dr. Eric Wright](#)

Resources and Services specific to State Waivers, Government Assistance, Insurance and Advocacy.

Angelman Family Contributions: Advocacy

People listen to you better when you are rational, realistic and appreciative of others' hard work. When you attack people in order to advocate for your angel, it never ends well for your angel.

AS Family Member

You are your own best advocate. Do not stop asking questions! Some of the doctors we now have are a neurologist, a behavioral therapist, a geneticist, a regular pediatrician, a gastroenterologist, a physical therapist, an occupational therapist, and a speech therapist. No one told us we should be seeing some of these doctors. We had to figure it out on our own. It really does take a village to raise a child with Angelman syndrome! Ask questions!

Elise

Be an advocate for your child in the community. If a "typical" child says or does something kind to your angel, praise the child by saying, "That is so kind of you. You are a wonderful boy/girl and (your angel's

name) really seems to really like you!" Other children will hear that and emulate this kindness. Use encounters with others as "teachable moments". Be a role model. Smile, make eye contact, and set the stage for others to approach your angel with ease.

Alice and Mark, sandiegasfwalk@gmail.com angel Whitney, age 38

Be the squeaky wheel. YOU ARE YOUR CHILDS VOICE!

Erin, erinctallant@gmail.com, angel, Gillie Mae, age 2.5

AFO

AFO is "ankle-foot orthotic". Orthotics are inserts for shoes that control abnormal foot function. They are used to realign the arch structure, realign in-toeing and out-toeing problems, and control the foot-ankle complex. AFOs can help improve mobility and balance. For further information speak with a physical therapist or orthopedist.

Angelman Family Contributions: AFO

My son was recommended for AFOs a few months ago. He's 2 (deletion +). They really do seem to help give him more support. Also I highly recommend ikiki shoes for young children using AFO's. There so much easier getting on and off feet.

Sarah j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

We struggled with shoes to fit over the AFOs and could never find anything that worked well. We finally bought a pair of Hatchbacks that are designed to fit over AFOs. They were more expensive but they lasted a lot longer and were safer for our daughter to wear.

Christine and Giovanni; Logan, Utah; angel Chiara

My son has to wear braces for the low tone in his ankles. The best AFOs we have found are SureSteps. They are cut in a way that allows my son to use the ball of his feet; they are not too rigid; and they fit in most regular shoes (with the inseam removed).

Michelle

Shoes that fit AFOs for kids: Plae, Billy's, and ikiki.

Tatiana Ortiz-Rubio, angel Alina, age 4, UBE3A Mutation

We have found that the "school uniform" socks in the girl's section at Walmart work wonders under SMO/AFOs. They come in packs of 3; white, black, & navy (so they are unisex)! They're thin, soft, breathable, very affordable, & they come up to the knee so that the AFO doesn't rub against any skin.

Shayla shaylacosborne@gmail.com, angel Dawson, age 7, Del+

ANGELMAN SYNDROME

Named after Dr. Harry Angelman, an English physician who discovered the syndrome, Angelman syndrome (AS) is a rare neuro-genetic disorder. It occurs in one in 15,000 live births. Angelman syndrome is often misdiagnosed as cerebral palsy or autism due to lack of awareness. Characteristic features of this condition include delayed development, intellectual disability, severe speech impairment, and problems with movement and balance (ataxia). Most affected children also have recurrent seizures (epilepsy) and a small head size (microcephaly). Delayed development becomes

noticeable by the age of 6 to 12 months, and other common signs and symptoms usually appear in early childhood. Children with Angelman syndrome typically have a happy, excitable demeanor with frequent smiling, laughter, and hand-flapping movements. Hyperactivity, a short attention span, and a fascination with water are common. Most affected children also have difficulty sleeping. Other features can include unusually fair skin with light-colored hair and an abnormal side-to-side curvature of the spine (scoliosis). The life expectancy of people with this condition appears to be nearly normal. Individuals with Angelman syndrome will require life-long care. Because of its genetic relationship to autism and other disorders, many researchers believe that curing Angelman syndrome will lead to cures for similar disorders. Angelman syndrome research is on the cusp of potential treatments to improve the debilitating symptoms of Angelman syndrome.

ANGELMAN SYNDROME FOUNDATION

www.angelman.org

See H- History to view a timeline: The History of the Angelman syndrome and the Angelman Syndrome Foundation.

See M-Marathon for information about this annual fundraising event.

See W- Walk for information about the annual ASF Walk fundraiser. <https://www.angelman.org/walk/>

Also, to view the history of Angelman syndrome and the Angelman Syndrome Foundation in a detailed and visual timeline format, visit: <https://www.angelman.org/about/history/>

Facebook: <https://www.facebook.com/AngelmanSyndromeFoundation>

Twitter: <https://twitter.com/angelman>

Instagram: https://www.instagram.com/angelman_asf/

Pinterest: <https://www.pinterest.com/angelmansyndfdn/>

YouTube: <https://www.youtube.com/user/AngelmanSyndromeFdn>

The mission of the Angelman Syndrome Foundation is to advance the awareness and treatment of Angelman syndrome through education and information, research, and support for individuals with Angelman syndrome, their families and other concerned parties. We exist to give all of them a reason to smile, with the ultimate goal of finding a cure.

The Angelman Syndrome Foundation has received a *4-Star rating from Charity Navigator* for demonstrating strong financial health and commitment to accountability and transparency. Four stars is Charity Navigator's highest possible rating and indicates that the ASF adheres to sector best practices, executes its mission in a financially efficient way and exceeds industry standards and outperforms most charities in our area of work.

ASF is a national 501(c)(3) organization dedicated to helping families, care providers and medical professionals arm themselves with as much helpful information about Angelman syndrome as possible. The ASF sponsors a biennial conference which gives you the opportunity to hear the latest research results, therapeutic techniques, educational strategies, long-term planning and financial-planning

information. The conference also offers many networking opportunities to talk to families that are dealing with the same issues you might be having while caring for an individual with AS.

The foundation sponsors AS research through grants to researchers who pursue promising avenues of discovery. Since 1996, the ASF has been funding research grants. See [ASF Funded Research](#) for more information.

The Angelman Syndrome Foundation (ASF) has invested more than \$9 million in Angelman syndrome research to date, supporting projects worldwide in the quest to find treatments for Angelman syndrome and ultimately a cure. Treatments resulting from the ASF's \$9 million investment in research help individuals with Angelman syndrome live better lives today and lead to better lives tomorrow, but require ongoing financial support.

The Angelman Syndrome Foundation is dedicated to funding the highest caliber of research on Angelman syndrome. It is our hope that these funded researchers, and their collaborators and peers, will bring forth new discoveries that ultimately lead to safe and effective therapies that improve the quality of life for all with Angelman syndrome.

More information from the ASF

The ASF's service population is individuals with Angelman syndrome and their families, care providers, service providers and other concerned individuals involved in the lives of people with AS. Angelman syndrome is a complex neurogenetic disorder that crosses all socioeconomic, race, and ethnic lines. It is estimated to occur in one in 15,000 live births, although this is thought to be grossly underestimated as there is a high rate of misdiagnosis in this population. The characteristics of Angelman syndrome include severe developmental delay, speech/communication difficulties, seizure disorders, significant behavioral issues and difficulties with movement, balance and walking. *As of today* there is no cure for Angelman syndrome and all individuals with AS will require life-long care and supports.

The Angelman Syndrome Foundation Board of Directors is made up by a diverse representation of backgrounds, skills and talents and at present, totals 14 members from across the United States. All but two directors have an individual with Angelman syndrome within their immediate family.

2019 ASF Staff

CEO:	AMANDA MOORE
DIRECTOR OF OPERATIONS:	EILEEN BRAUN
SPECIAL EVENTS COORDINATOR	KITTY MURPHY
CLINICAL DIRECTOR:	ELIZABETH JALAZO
WEBMASTER:	AMY FRIEL

2019 ASF Board of Directors:

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Director: Anna Blanding
Director: Rebecca Burdine
Director: Leslie McCallister
Director: Susan Ravellete
Director: Jim Lamb

ASF Programs, Services and Supports

The Angelman Syndrome Foundation provides information and resources so that families of individuals with Angelman syndrome can better manage the ages, stages and special needs of those with the syndrome throughout their lifespan. This is accomplished through a variety of programs, centers of excellence, educational opportunities, special events and research.

Core Programs

Angelman Syndrome Behavioral Series <https://angelmanbehaviors.org>

This web-based training series was developed to help families deal with the daily challenging and often aggressive behaviors that people with Angelman syndrome exhibit. Prepared by a panel of clinical experts in AS (including psychology, psychiatry, neurology, epilepsy and communication) the series provides access to invaluable help and behavioral information from any device, at any time, any place and at no cost to the user.

ASF Educational Webinar Series and Communication Training Series

<https://www.angelman.org/resources-education/educational-webinars/>

<https://www.angelman.org/resources-education/communication-training-series/>

Webinars by AS experts on AS-specific topics, including but not limited to research updates, clinical developments and help for daily living and management of AS is available and accessible to all AS families 24/7/365 and around the world.

The Communication Training Series was developed to break down often very complex communication training and instruction into smaller, simpler parts, where care providers and their support teams can go at their own pace as they are working with their special person with AS. The series includes training tools and how to use picture symbols, includes downloadable picture and symbol display templates and step-by-step instructions to start and continue a communication program at home and school, no matter the individual's age, skill level or location. The ASF Communication Series was developed by a leading team of experienced communication experts in AS and other non-verbal populations.

To date, the ASF educational webinar and communication training series have collectively seen over 75,000 views. The series continue to be used extensively throughout the global AS community and a future goal is to translate the ASF Communication Series into Spanish.

ASF Family Resource Team <https://www.angelman.org/resources-education/asf-family-resource-team/>

An invaluable resource to AS families, the *ASF Family Resource Team* is a group of compassionate AS experts who have a child or family member with AS. These individuals serve as a lifeline for AS families, providing guidance and understanding on topics such as health, insurance, finance, education, transportation, technology, durable medical goods and equipment and much more. Resources at a local, state and federal level are provided to AS families across the US.

ASF AS Clinic Network <https://www.angelman.org/angelman-syndrome-clinics/>

The Angelman Syndrome Foundation founded the AS Clinics to provide individuals with Angelman syndrome the specialized, comprehensive medical care they need, from birth through adulthood. The ASF has partnered with world-class, leading medical and research institutions in the US and internationally to establish AS Clinics to provide comprehensive medical care individuals with AS need, all in one location. Each clinic has its own unique capabilities that leverage a variety of expertise and specialized care available from each partnering organization.

Additionally, all clinics in the ASF AS Clinic Network are working together to establish standards of care for treatment of AS. Clinicians often consult with other clinicians at other network clinics and work together to find the best possible outcomes for their patients.

Beyond specialized medical care, the ASF currently funds—and will continue to fund—research that occurs at AS clinic sites. Data gathered from the AS Clinics is, and will continue to be, used to drive research for viable treatments and a cure for AS. ***The ASF AS Clinic network is THE only clinical framework established to support future clinical trials when they become available by having established sites with AS experts and patients in place.*** This has always been a part of the ASF research strategic roadmap. The ASF AS Clinic Network is intentional in its abilities to provide high-quality clinical care today, across the AS lifespan, as well as be THE platform for AS clinic trials.

Education – Annual Scientific Symposium and Biennial Conference

<https://www.angelman.org/conferences-symposia/>

Since 1991, the Angelman Syndrome Foundation has sponsored 15 family educational conferences and 18 scientific/research symposia that bring together the foremost authorities on Angelman syndrome.

The world’s premier research symposia are the proverbial “think tank” that attracts researchers from around the globe who gather and share findings on the latest advances in the field of Angelman syndrome. Rarely seen in scientific meetings, and similar to Gordon-style meetings, the ASF research symposia bring the brightest and best researchers together to share their *unpublished data* with others in a *collaborative environment* to push research toward viable treatments and cure forward at a quicker pace.

The biennial family conference provides an opportunity for parents and families with an individual with Angelman syndrome to come together to hear and learn about the latest research findings, development in treatments, therapies and management of Angelman syndrome. It also offers time to share and networking opportunities for families who are newly diagnosed to veteran care providers.

Investment in Research <https://www.angelman.org/research/asf-funded-research/>

Just as the Angelman Syndrome Foundation is there to support those living with Angelman syndrome, it also envisions a future in which individuals with Angelman syndrome and their families will no longer be affected by the myriad of medical, behavioral, emotional, financial and practical issues that come with Angelman syndrome.

The ASF prides itself on funding the highest-quality research studies from the best AS research institutions, studies that have long-term results to improve the quality of life for people with AS, leading us closer to treatments and a cure.

Since 1996, 90 research grants have been awarded by the Angelman Syndrome Foundation (as of this printing), totaling over \$9.5MM dedicated to research toward therapeutics and a cure. These high-quality research results have leveraged over \$25MM additional expanded studies with the NIH, the DOD, the Simon's Foundation and other funding sources. Over 100 research studies with researchers spanning the US and 10 countries across the globe have been funded. In short, the ASF does not limit its funding to a small group of researchers but seeks out the brightest and best to cure Angelman syndrome.

Angelman Family Contribution: ASF

The Angelman Syndrome Foundation is a great resource for families! Especially those that have a child recently diagnosed! It was so reassuring to see so much informative information about Angelman syndrome! I gained so much knowledge from the AS walk and Family conference in Louisville, KY! This is a great place to start! Amazing individuals who truly care!

AS Family Member

AS CLINICS

<https://www.angelman.org/angelman-syndrome-clinics/>

By partnering with leading medical and research institutions across the country, the ASF founded the Angelman syndrome clinics to provide individuals with AS, from birth through adulthood, the comprehensive medical care they need throughout their lifetime. Each Angelman syndrome clinic is completely comprehensive yet has its own unique capabilities that leverage the expertise and specialized care available from each partnering organization. At the clinics, individuals have access to a variety of professionals all specializing in AS:

- Clinical geneticist
- Neurologist
- Psychiatrist
- Psychologist
- Speech Language Pathologist
- Physical/Occupational Therapist
- Genetic counselor
- Social worker
- Nutritionist

Current AS Clinics include:

- Buenos Aires, Argentina
- Children's Hospital Colorado, Aurora, Colorado *** *Opening in 2020*
- Children's Hospital of Eastern Ontario
- Edmond and Lily Safra Children's Hospital, Tel Hashomer
- Erasmus MC, Rotterdam
- Geisinger Janet Weis Children's Hospital, Danville, PA
- Massachusetts General Hospital
- Mayo Clinic
- Minnesota Epilepsy Group
- Nicklaus Children's Hospital, Miami
- NYU Langone Health
- Rady Children's Hospital San Diego

Rush Univ Medical Center, Chicago
Seattle Children's Hospital
Texas Children's Hospital, Houston
UCLA Cart
UCSF Benioff Children's Hospital, Oakland
University of North Carolina, Chapel Hill
Vanderbilt Univ Medical Center, Nashville
Weill Cornell Medicine, New York

The Angelman syndrome clinics also provide a foundation to support future clinical trials by having established sites with AS experts and patients in place to conduct those trials when they become available.

Angelman Family Contribution: ASF Clinics

We see Dr Thibert in the Angelman clinic at Boston Mass General. He is one of the most knowledgeable physicians when it comes to AS.

AS Family Member

ANGELMAN TODAY

ANGELMAN TODAY – A GLOBAL PERSPECTIVE ON ANGELMAN SYNDROME

Lizzie Sordia, Editor in Chief and Publisher of Angelman Today

Angelman Today is an online resource featuring information, research, and inspiration for all things related to Angelman syndrome. It is the first and only publication of its kind. Created for families by families. Our articles feature information that we hope will revolutionize the current care and standard of practice for the treatment and education of those affected by AS.

Angelman Today offers every Angelman syndrome organization across the globe the opportunity to submit articles so that everyone can benefit from the brilliant minds involved in the care, research and treatment of our loved ones with AS. We embarked on this project to put the best and brightest from around the world at your fingertips.

Angelman Today is a lifestyle guide to achieving better health for individuals with Angelman syndrome and their families. It is written with your needs in mind but is not a substitute for consulting with your physician or other health care providers. The publisher and authors are not responsible for any adverse effects or consequences resulting from the use of the suggestions, products or procedures that appear on this website or online magazine. All matters regarding your health should be supervised by a licensed health care physician.

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ANIMALS

Angelman Family Contributions: Animals

Our family now has a Goldendoodle for our daughter Maddy, age 12. We will be training him for a service dog. He is very smart, aware, and treats her very differently than the rest of the family. This is the perfect temperament for a service dog. He only licks and kisses her and does not attempt to jump on her or play rough with her. She has learned how to be gentle and kind from the dog.

Myriah, angel Madeline, age 12, Mutation

My son Brantley is 2 and deletion+ and loves animals! We have a house dog "Molly" that adores him! I find that he is always so interested in animals. Also equine therapy is awesome for children with AS!
Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

ANXIETY

<https://onlinelibrary.wiley.com/doi/full/10.1111/jir.12635>

Anxiety-associated and separation distress-associated behaviors in Angelman syndrome

A. C. Wheeler K. C. Okoniewski A. Wylie M. DeRamus L. S. Hiruma D. Toth R. B. Christian

First published: 28 May 2019

Background

Anxiety is considered a 'frequent' feature in the clinical criteria for Angelman syndrome; however, the nature and severity of anxiety symptoms have not been well characterized in this population. Anxiety behaviors, especially in response to separation from a preferred caregiver, have been described clinically but have not yet been explored empirically.

Method

This study used a combination of standardized and clinician-derived survey items to assess the frequency, nature and severity of behaviors associated with anxiety and separation distress in 100 individuals with Angelman syndrome. Family (e.g. income and maternal education) and individual (e.g. age, sex, genetic subtype, sleep difficulties and aggressive behaviors) variables were also gathered to assess possible predictors of higher anxiety levels. Approximately half of the sample was seen in clinic and assessed with standardized measures of development and daily functioning, allowing for an additional exploration of the association between anxiety symptoms and extent of cognitive impairment.

Results

Anxiety concerns were reported in 40% of the sample, almost 70% were reported to have a preferred caregiver and over half displayed distress when separated from that caregiver. Individuals with the deletion subtype and individuals who are younger were less likely to have anxiety behaviors. Sleep difficulties and aggressive behavior consistently significantly predicted total anxiety, the latter suggesting a need for future studies to tease apart differences between anxiety and aggression or anger in this population.

Conclusions

Anxiety concerns, especially separation distress, are common in individuals with Angelman syndrome and represent an area of unmet need for this population.

***There are several other limitations to this study, which include lack of comprehensive, diagnostic measures of anxiety; the cross-sectional nature of the study; and the lack of information on several potentially important key variables, such as functional communication skills, co-morbid conditions such as seizures, pain or gastrointestinal issues, medication use and behavior management techniques used by caregivers. However, **anxiety-related symptoms are a common and challenging aspect of the AS phenotype, one that deserves greater attention.** This paper provides additional evidence of the frequency, severity and nature of behaviors thought to be related to anxiety in this population. **Being able to identify better both the molecular and environmental contributions to these behaviors is an important goal for researchers and clinicians working to improve the quality of life of individuals with AS and their families. Future comprehensive studies are needed to better capture symptoms of anxiety in individuals with AS over time to better understand this associated feature.**

Angelman Family Contributions: Anxiety

As our son has gotten older, anxiety has become a huge part of his life. He is very attached to his familiar caregivers. When he wants something that he cannot have, he has behavioral outbursts because he cannot communicate his wants and needs. Buspar is the medication that has helped the most with this.
AS Family Member

Cuddle swings and similarly enclosed forts/supportive chairs (like bean bags) help our child to calm himself because it gives his body more awareness and grounding, helping him to have more control as he knows where he is at in space.

Desiree, desireemartika@yahoo.com, angel Titus, age 3, Del+

ASF FAMILY FUND

<https://www.angelman.org/resources-education/asf-family-fund/>

The ASF Family Fund was created to assist families supporting individuals with Angelman syndrome. The ASF Family Fund allows families to apply for funds that would allow access to resources that are needed to improve the quality of life for an individual with Angelman syndrome.

Some possible resources that could be funded in part or in whole through the ASF Family Fund are:

- Support to access an ASF Angelman Syndrome Clinic. This could include travel to the nearest clinic to your current location.
- Support to access safety support (safety bed, stroller, etc).
- Support for communication devices.
- Support for purchase of adaptive play (adaptive bike, etc).

To complete an application:

<https://www.angelman.org/resources-education/asf-family-fund/>

ATAXIA

Ataxia is a neurological sign consisting of lack of voluntary coordination of muscle movements that can include gait abnormality, speech changes, and abnormalities in eye movements. Ataxia is a clinical manifestation indicating dysfunction of the parts of the nervous system that coordinate movement, such as the cerebellum.

Children with Angelman syndrome may hold their arms up with the wrists and elbows bent and may flap their hands repeatedly when walking or excited. Diminished muscle tone (hypotonia) of the trunk, increased muscle tone (hypertonia) of the arms and legs, and abnormally exaggerated or brisk reflex responses (hyperreflexia) may also occur. Some children with Angelman syndrome experience subtle tremors of the arms and legs. These movement disorders may be apparent early during infancy (approximately 6-12 months of age). Motor milestones (e.g., walking) are usually delayed. In mild cases, children may begin to walk at 2-3 years of age. In more severe cases, walking may be noticeably slow, stiff and jerky. Some children may not be able to walk until they are 5-10 years of age. In approximately 10 percent of cases, children with Angelman syndrome do not walk unaided.

Angelman Family Contribution: Ataxia

Our son is extremely mobile but does have ataxia. Shoes with good support help, and making sure there are not things lying around that he could easily trip on.

AS Family Member

AUDREY ANGELMAN

Biographical Information by Mrs. Pam Kempt, niece of Audrey Angelman 1999

Pam is the daughter of Jean Walsh, Audrey's older sister



Audrey Stuart Angelman was the younger daughter of Mr. and Mrs. A.S. Taylor. Audrey was educated at Merchant Taylor's School for Girls Crosby, Liverpool. During her time at school she was junior tennis champion and won two awards for public speaking. Audrey was awarded scholarships and attended Liverpool University where she achieved BA and B Ed degrees. Audrey was an accomplished pianist, organist and writer.

She became Deputy Head of the King David School, Liverpool where she was highly respected. Audrey wrote and composed the school song and formed the school choir. Audrey later left the King David School to take a position as a lecturer at St. Katherine's College, Liverpool. In December 1964 Audrey married Dr. Harry Angelman who was a pediatrician at Warrington Hospital. At the time, Harry was working on identifying the very early stages of a condition that would later be named for him, Angelman syndrome. After Harry's passing, Audrey worked very hard for the Angelman Syndrome Foundation and successfully established her dream of an International Angelman syndrome Organization. Through her travels she met many children with Angelman syndrome along with their parents and relatives. Audrey kept all the letters she received and made a point of personally replying to each one.

Sadly, Audrey passed on August 15, 1999 in England after a brief illness. She faced this illness with her usual stoicism and wry sense of humor. That autumn, the Angelman Syndrome Foundation announced the creation of the "Harry and Audrey Angelman Award" in their honor. The award recognizes an

individual or group who have demonstrated a strong commitment to enhancing the awareness and understanding of Angelman syndrome.

Tribute by Dr. Joe Wagstaff, April 2000

Audrey Angelman felt that children with disabilities and their parents suffer as much from isolation and lack of communication as they do from the disabilities themselves. One of her great gifts was for communication, and she put an enormous amount of time and effort into trying to end that isolation for AS families. She felt that communication between AS families provided the great emotional relief of knowing that someone else had been through the same experiences and joys and heartbreaks. She also knew that mothers, fathers, brothers and sisters, grandmothers, grandfathers, and cousins of AS individuals find solutions to problems that would never occur to therapists or physicians. Angelman syndrome from A to Z is a perfect expression of Audrey's desire to bring AS individuals and their families together, to share solutions to problems where they exist, and to provide support and comfort when the problem hasn't been solved yet.

Dr. Joe Wagstaff (1955-2008) Audrey considered Joe "like a son". Dr. Harry and Audrey Angelman never had children of their own.

Anecdote *Alice Evans, mom of angel Whitney, age 38*

In 1997 Audrey stayed with us for a few days in our San Diego home. It was the year after Harry's passing and immediately after the 1997 ASF Seattle Conference. It was very important for those who planned the Seattle conference to make sure that Audrey felt she was still a vital part of the ASF and the Angelman community. Audrey was scheduled to host the "Ask Audrey" session, and I sensed beforehand that Audrey was worried no one would want to attend. I can't tell you how pleased she was when the entire room began to fill!! Of course, she included Harry in every response she gave no matter how specific the question was about her.

While she was at our home, I taught Audrey how to use email. I asked members of the Angelman Listserv to email Audrey on that particular day. Imagine her surprise when dozens and dozens of emails began popping up! She was so thrilled and we printed each one. At breakfast the next morning she had the huge stack of printed emails in her hand. Audrey confessed that she had been up all night reading them over and over again! Audrey and Harry never had children of their own, but they most certainly considered all of the angels and their families to be "theirs".

DR. HARRY ANGELMAN *August 13, 1915- August 8, 1996*



Dr. Harry Angelman, an English physician at Warrington General Hospital, published a research paper that first described children with characteristics of Angelman syndrome. At that time, he referred to the disorder as "Happy Puppet Syndrome." The condition was considered to be extremely rare at that time, and many physicians doubted its existence. This discovery, in theory, was the birth of AS.

Read his paper, *Puppet Children, a Report on Three Cases:*
Developmental Medicine Child Neurology 1965 681-688

https://onlinelibrary.wiley.com/doi/epdf/10.1111/j.1469-8749.1965.tb07844.x?purchase_site_license=LICENSE_DENIED&show_checkout=1&r3_referer=wol&purchase_referrer=en.wikipedia.org&tracking_action=preview_click&

Dr. Angelman was born in Birkenhead in 1915 and qualified in Liverpool. Angelman was an enthusiast for the language and country of Italy. He had observed three children who were unrelated but showed similar symptoms (severe intellectual disablement, lack of speech, motor disorders, and happy demeanors). He was of two minds as to whether he should publish his findings but he described seeing a painting which seemed to characterize the symptoms he had observed. The painting showed a boy with a puppet and was by the renaissance artist Giovanni Francesco Caroto. Angelman travelled to talk about his discovery and his work was mentioned as important by U.S. President Bill Clinton. Angelman died due to a colon tumor.

https://en.wikipedia.org/wiki/Harry_Angelman

AUTISM-LIKE FEATURES

The following information is taken from 2009 document written by Charles A. Williams, M.D; and Saria U. Peters, Ph.D.

[http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20as%2009%203-19-10.pdf)

Some of the associated clinical features of AS (e.g. hand--flapping, stereotypic behaviors, deficits in expressive language), overlap with certain features of autism. Generally speaking, clinicians should exercise caution when examining symptoms of autism within AS, because some AS patients have been mistakenly identified as having autism in lieu of AS [41], and some patients who exhibit features of autism when they are younger, may no longer exhibit these features as their cognition and their language skills improve.

There are, however, some studies that specifically examine the frequency and magnitude of autistic traits in individuals with AS. While some researchers demonstrate a lack of autistic traits or very low incidence of autism in individuals with AS [42, 43], several other studies have demonstrated that a percentage of individuals with AS do also meet criteria for autism [34, 35, 44]. Individuals with AS and co--morbid autism are more likely to show decreased eye gaze, fewer social overtures, use fewer nonverbal gestures, use another person's body as a "tool" to communicate "for" them, have decreased shared enjoyment in interactions, and fewer socially directed vocalizations [34, 44].

Recent studies demonstrate that it is primarily deletion positive individuals with AS that exhibit greater symptom severity associated with autism, and within the deletion positive group, primarily children with larger, Class 1 deletions [45] [46]. Most recent findings indicate that these differences in symptoms of autism between the deletion subgroups are not related to differences in cognition (i.e. children with greater symptom severity were not necessarily lower functioning).

To summarize, studies seem to indicate that severity of autism symptoms in AS only affects a small proportion of AS patients, is associated with deletion size, and with a more aloof/withdrawn behavioral phenotype. There are four genes (NIPA 1, NIPA 2, CYFIP1, & GCP5) missing in Class I and present in Class II deletions (refer to diagram in Genetic Mechanisms that Cause AS section), one or more of which may have a role in the development of socialization skills and symptoms related to autism. For the small

percentage of patients with AS who do exhibit more features of co-morbid autism, specific therapies such as applied behavioral analysis are quite helpful.

Longitudinal follow-up of autism spectrum features and sensory behaviors in Angelman syndrome by deletion class

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sarika.u.peters@vanderbilt.edu

<https://www.ncbi.nlm.nih.gov/pubmed/?term=21831244>

Conclusions: Despite a lack of differences in cognition or adaptive behavior, individuals with Class I deletions have greater severity in ASD features and sensory aversions that remain over time. There are four genes (NIPA 1, NIPA 2, CYFIP1, and GCP5) missing in Class I and present in Class II deletions, one or more of which may have a role in modifying the severity of social affect impairment, and level of restricted/repetitive behaviors in AS. Our results also suggest the utility of a dimensional, longitudinal approach to the assessment of ASD features in populations of individuals who are low functioning.

Zylka Lab awarded \$6.1 million from NIH to develop CRISPR/Cas9 gene therapy for Angelman syndrome, study UBE3A autism gene with Philpot Lab

<http://news.unhealthcare.org/news/2019/september/zylka-lab-awarded-6-1-million-from-nih-to-develop-crispr-cas9-gene-therapy-for-angelman-syndrome-study-UBE3A-autism-gene-with-philpot-lab>
September 17, 2019

The National Institutes of Health have awarded two separate grants totaling \$6.1 million to Mark Zylka, PhD, director of the UNC Neuroscience Center. One of the grants was co-awarded to Ben Philpot, PhD, associate director of the center at the UNC School of Medicine.

One of these projects will test a CRISPR/Cas9-based gene therapy for Angelman syndrome in mice and human neurons. Zylka is the principal investigator for this project, which is funded by a \$2.8-million grant from the National Institute of Neurological Disorders and Stroke.

When the maternal copy of the gene UBE3A does not work properly, the result is Angelman syndrome. The paternal copy is normally turned off, or silenced, but has the potential to serve as a backup for the faulty maternal copy. Zylka's lab is using gene editing to unsilence the dormant paternal copy of UBE3A. "There is currently no effective treatment or cure for Angelman syndrome," said Zylka, the W.R. Kenan Distinguished Professor of Cell Biology and Physiology. "Our research will provide the first preclinical evidence that CRISPR/Cas9 can be used to enduringly 'unsilence' the paternal UBE3A gene in mice and 'unsilence' paternal UBE3A in cultured human neurons. This new knowledge has the potential to advance a first-of-its kind treatment for a pediatric-onset autism spectrum disorder."

The second grant builds on research published in Cell by the Zylka lab, and seeks to better understand how a genetic mutation in UBE3A contributes to certain characteristics of autism. Zylka and Ben Philpot, PhD, Kenan Distinguished Professor of Cell Biology and Physiology, are co-principal investigators for this project, which is funded by a \$3.3-million grant from the National Institute of Mental Health. "Our research will evaluate the extent to which UBE3A gain-of-function contributes to progenitor cell proliferation, proteasome dysfunction, and other autism-related phenotypes. This new knowledge could point towards a new therapeutic strategy for autism – one based on rebalancing UBE3A and proteasome function in the developing brain," Zylka said.

Zylka was initially awarded financial support through the Angelman Syndrome Foundation for proof-of-principle studies with CRISPR/Cas9. And earlier this year, the Zylka lab was awarded \$6.8 million to investigate interactions between genetics and environmental exposures that may contribute to neurodevelopmental disorders such as autism and attention deficit disorder.

Angelman Syndrome's Silent Gene Points Way Forward for Autism Therapies

By Stormy Chamberlain / 21 OCTOBER 2019

<https://www.spectrumnews.org/opinion/viewpoint/angelman-syndromes-silent-gene-points-way-forward-for-autism-therapies/>

Stormy Chamberlain

Associate professor of genetics and genome sciences, University of Connecticut

Stormy Chamberlain is associate professor of genetics and genome sciences and associate director of the Graduate Program in Genetics and Developmental Biology at the University of Connecticut, School of Medicine.

The autism community should be paying close attention to Angelman syndrome, a condition related to autism that is caused by mutations in a single gene.

Investments in basic and clinical research, along with the efforts of an intrepid community of families, has brought us to the brink of a 'cure' for the syndrome. If successful, these efforts could provide a blueprint for treating other conditions, including autism.

Angelman syndrome is characterized by a range of features, including developmental delay, motor problems, seizures, sleep problems and, in many cases, autism. Most people with the syndrome are minimally verbal, walk with a wide-spaced, unsteady gait (ataxia) and show repetitive behaviors such as hand-flapping. Despite these difficulties, they are frequently exuberant and happy.

The gene mutated in the syndrome, UBE3A, holds the key to potential therapies. Only the copy of UBE3A inherited from the mother is typically active in neurons. In people with Angelman syndrome, this maternal copy is nonfunctional; the paternal copy is perfectly good — but silent.

Treatment strategies center on restoring expression of UBE3A by either replacing the maternal copy — gene therapy — or activating the silent paternal copy. Scientists have made progress on both fronts.

Gene therapy

Directly replacing the gene may seem like the most straightforward approach. Viruses can deliver a gene as small as UBE3A to neurons efficiently. The challenge is to regulate how many copies of the gene are delivered to cells and how much protein is produced from each copy — because having too much of the protein can also cause problems.

Because of this, multiple academic groups and pharmaceutical companies are trying to carefully calibrate the gene dose as they develop gene therapies for Angelman syndrome. Among the teams at work on gene therapy for Angelman syndrome are those at the University Of Texas Southwestern Medical Center, the University of Pennsylvania and PTC Therapeutics.

Another challenge is that, in people, the UBE3A gene can produce three slightly different proteins located in different parts of a neuron. One form is predominant, but it is not clear whether the other forms have specific roles. Gene therapy can likely replace only one form. So if the different forms of

UBE3A have different functions, gene therapy may only be able to restore the function of one of the forms.

The second approach, activating the silent paternal copy of the gene, also seems feasible. In 2011, researchers established proof of principle of this approach. They screened about 2,400 small molecules to see if any of them could turn on the silent copy¹.

One that did is a topoisomerase inhibitor, a blocker of enzymes that untangle DNA. Inhibiting these enzymes may halt the production of an RNA that silences UBE3A. However, topoisomerase inhibitors have side effects, because they influence other genes, too.

Small DNA-based drugs can do the same thing with more precision. These drugs are similar to nusinersen (marketed as Spinraza), a treatment for spinal muscular atrophy. Nusinersen is an antisense oligonucleotide that boosts the expression of a vital motor neuron gene.

Making antisense

A few years ago, researchers tested an antisense oligonucleotide for Angelman syndrome. The molecule unsilenced paternal UBE3A and improved cognition in a mouse model². Based on this work, three companies (Ionis, Roche and GeneTx) are pursuing similar therapies for the syndrome. These drugs are likely to require repeated spinal injections.

Another tactic involves CRISPR/CAS9, a programmable DNA editing system. Though the system works like topoisomerase inhibitors do, it is probably far more precise. Importantly, it can be packaged into a virus and delivered like other gene therapies. A range of RNA-cutting molecules can also be packaged into viruses to deliver the drug in a one-and-done fashion. Yet other methods include small molecules that activate paternal UBE3A by unknown mechanisms.

Clinical trials of some of these therapies could begin within a year or two. It is exciting to have multiple 'shots on goal' for Angelman syndrome.

Researchers and advocacy groups are discussing newborn screening and prenatal diagnosis so that everyone with Angelman syndrome has access to these therapies when they might be most effective. This Herculean effort would not be possible without the collaborative efforts of all corners of the community.

If we can develop a transformational therapeutic for Angelman syndrome — and it seems almost certain we will — it is likely to pave the way toward similar solutions for autism.

To quote Booker T. Washington, "Success always leaves footprints."

Angelman Family Contribution: Autism

More than twenty-five years ago I had a student in my classroom with autism. Even though he was quite different than my angel, I did notice some similar features, such as: arm flapping; hyperactivity; difficulty altering routines; communication challenges; sleeping problems (as reported by his parents); attention issues; and more. I was told repeatedly that there was no connection between Angelman syndrome and autism. However, I was convinced that there just might be a link. (This was a time when immunizations, along with other theories, were suspected as being a cause for autism.) My daughter is Deletion Positive and now it is thought that children who have a large deletion like her, labeled Class 1, do have some

features of autism. My take away from this is that we parents do have some unique insights when it comes to our children. Keep speaking up! You might be on to something!

Alice, sandiegoasfwalk@gmail.com, San Diego, CA, angel Whitney, age 38, Del+ Class 1

B

BATHING

Angelman Family Contributions: Bathing

Do not rush bath time. Be consistent and have a routine...

lather/ rinse, break, shampoo, rinse, break, conditioner, rinse, break.

Breaks aren't long, maybe 2min. Let them know you are done and she will need to be out of the tub in 2 minutes.

Norma, Murrieta, CA, angel Katarina, age 20, Del +



A walk-in shower and shower chair have made bathing so much easier.

Jo Lynn and Jeff, angel Caitlynn, UPD

We recently got a bath seat that holds approximately 250 lbs. I put it in the tub. When my son sits on it, there is a remote that lowers him in the tub. I make it a game and sing going down-down-down! After I wash him, I sing going up-up-up! This makes it easy on your back.

Aina, Fairfield, Ohio, angel Ra'Shawn

Bath time seems to be the highlight of the day! Parents capitalize on this opportunity to take care of a few of the more difficult jobs ...like haircutting and nail clipping! Since getting the child OUT of the tub (willingly) is a huge feat, one parent has come up with a highly effective motivator! She warms the bath towel in the dryer for a few minutes, and the child has learned to look forward to that treat after her bath!

AS Family Member

Our angel spent most of bath time swatting at us or trying to lick the tub faucet. We turned bath time into "disco bath time party!" We blast his favorite songs, turn off the lights and turn on a colored light show, and I also throw a couple glow sticks in the tub for good measure. He LOVES it!! He sits there and dances or plays with the glow sticks while I scrub him and wash his hair without a struggle.

AS Family Member, angel age 7, UPD

Our angel has always enjoyed a bath. We find showering too difficult due to balance and safety concerns, but we do practice before camp so that he knows what to expect. We recommend keeping bottles of shampoo, body wash, etc. out of reach and sight as these are tempting! Also have durable bath toys like foam letters, shapes, ducks, etc. As they get older, they can help rub soap and shampoo on, pour water, and dry self.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

BEDS & BEDROOMS, PALJAMAS, WEIGHTED BLANKETS

Beds

Safe Place Bedding

Portable, inflatable beds provide a safe sleeping solution for special needs loved ones at home or away. Washable mattress made of non-synthetic material. (Most mattress pads are made from synthetic materials that can raise the body temperature.)

The Safety Sleeper

The tent-like structure is durable, portable and lightweight (under 25 lb.) with additional safety features such as zipper locking mechanisms, enclosed mattress to protect against entrapment, fire retardant material, stabilizing straps to prevent tipping, and frame pads.

Cyr Designs, LLC

The Courtney Bed is an alternative to a standard hospital bed and is hand-crafted from solid hardwood, there is no metal, PVC or any other material used in fabrication.

Enclosed Crib/Bed Design

Enclosed bed for a growing child with a gate at the end allow parents easy access without a “cage” feel or look. Contact Bill Bonnell at wtbonnell@yahoo.com or the Cutolo’s at jjcutolo14@verizon.net.

<https://safesleepsystems.com>

The Nickel Bed Tents are designed to fit standard twin-sized mattresses. The mattress fits inside the base of the tent and helps keep children with disabilities from leaving their bed throughout the night.

Olivers Bed

Available in the **UK only**. Designed and build by a cabinet maker and AS parent.
Email: Info@oliversbed.co.uk

Pajamas

Pajama City

Find adult size footed pajamas.

Jumpin Jammerz

Find adult size footed pajamas.

Little Keeper Sleepers

A sleeper that is easy for caregivers to get on and off, but not one the children can get out of. Children stay warm at night and ensure no mess to clean in their beds in the middle of the night or come morning.

Weighted Blankets

Stitches by Anne <https://www.stitchesbyanne.info/>

Stitches by Anne makes quality weighted blankets, lap pads, vests, hats, shoulder wraps, bibs (baby to adult), cooling vests, and Animals “Critters” (to drape around your neck) at very affordable prices. Weighted products provide deep pressure touch stimulation providing a naturally calming effect.

<https://www.amazon.com/weighted-blankets/s?k=weighted+blankets> Amazon.com

Angelman Family Contributions: Beds and Bedroom

I wish someone would have told me to transition our son to an enclosed safe bed as soon as he was too big for a crib. No amount of sleep training or sleep meds have resulted in our son easily falling asleep and consistently staying asleep for at least 8 hours a night. Having a safe bed for him means that he can be awake at night and we know that he is safe. It allows the rest of our family to get the much needed rest that they need. *Abrams Safety Sleeper* is a great travel bed option because our kids' sleep patterns are really affected by change. For years we didn't travel because we had no safe place for him to sleep.
AS Family Member

[Our son] went from a crib right to a safe bed at age four. Insurance wouldn't cover a safe bed, so my husband built one for him. We opted for a double bed, which was a great choice because [he] moves around a lot. Three sides are solid wood and the fourth side is a gate that [he] can see through, we can hold his hand and not be in bed with him. We can also see him clearly on the video monitor that is mounted to his wall. [Our son] is 9 years old and loves his bed. He often goes there during the day if he's overwhelmed and needs a break.

AS Family Member

My daughter's room is one of her favorite spaces. We keep it simply decorated with only a swing and bed/sleeping tent. She also has a place for all her favorite, safe toys in a small bin. We want her space to be hers, conducive for safe play, good sleep, and afternoon therapy sessions. We have loved the Privacy Pop for our daughter. We can take this with us on trips, as well, so she feels right at home wherever we travel.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

When my angel was little, we designed her bedroom around her specific needs. Her mattress and box spring were placed directly on the floor so she couldn't climb under them. We installed a Dutch door with a latch on the outside so she couldn't wander at night. With the door we could still hear her if she cried, and she could see and hear her sisters and brother in their rooms. Instead of a dresser, we bought a kitchen cabinet and installed it on the wall of her room so she couldn't dump her clothes on the floor. She had only stuffed animals and soft toys in her room. Any plastic or metal toys were kept in the family room so she couldn't throw them around and hurt herself or damage anything in the room.

Lori, angel Adrienne

Hang bedroom drapes or curtains with Velcro. Our son is likely to pull them down, so this makes it easy to put them back up!

John, Travelers Rest, SC, angel Tipton, age 49

Use air mattresses around the bed in the bedroom. It keeps the carpet cleaner and the air mattress can be taken out and hosed off.

John, Travelers Rest, SC, angel Tipton

Margaux sleeps in a made-to-measure sleeping bag (adult sizes available) to avoid access to nappy.
www.ptitifilou.com

Estelle, Singapore, angel Margaux, age 14

Invest in water proof mattress pads for all beds in the home and be sure to have several sets of clean sheets and blankets on hand at all times!

Kathleen

Using a baby monitor at night helps alert to seizures or getting sick from reflux.
AS Family Member

My angel has slept through the night since she was seven. We kept her room really bland. All she had in there were her bed and a wardrobe. There were no pictures on the walls. Nothing hanging down, no light shows. We gave her no reason to want to be awake and it's paid off. I know it'll seem hard to give your child a 'dull' room, but sleep is vital for our angels' development and your sanity! Keep toys in a different room and you will give him the message that this room is for play, but when he goes to bed, it's to sleep. Good luck!

Kathryn, angel Darcy

When younger, especially, it helped to put the mattress on box springs directly on the floor in case of rolling out of bed, and putting soft cushiony items around the bed. We also learned to only put minimal décor in the room (baby proof for life??)! We used baby monitors for a long time! We let our son chew a cloth to settle in. We close his door so that we can hear if he leaves his room when he opens the door.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

The Safety Sleeper by Abrams Nation has been a lifesaver for us when traveling and in emergency situations. Paul loves the enclosed feeling and after trying another brand, this one holds up to the strength of our son. It is so well made and comes in its own suitcase (which is not charged as luggage when flying. We say it is a medically necessity).

Karen, wbk_sd@yahoo.com, angel Paul, age 15, Del+

BEHAVIOR

Additional information can be found at the ASF Behaviors Informational Series:

<https://www.angelman.org/what-is-as/angelman-syndrome-behaviors-informational-series/>

The most frequent requests for guidance and assistance from families of individuals with Angelman syndrome are related to behaviors. You are not alone! The Angelman syndrome Behaviors Informational Series is available to help those who care for or work with individuals with AS explore the possible root causes of challenging and aggressive behaviors and identify possible solutions.

Using text and corresponding videos that feature world-renowned experts on Angelman syndrome, the Angelman syndrome Behaviors informational series is made up of interactive modules. Each module addresses a particular aspect of behavior including:

1. Social and Environmental Influences on Aggressive Behavior
2. Mental Health Influences on Aggressive Behavior
3. Cognitive Issues and Sensory Impairments
4. Neurologic and Medical Influences on Aggressive Behavior
5. Social and Environmental Influences on Aggressive Behavior

GET STARTED

1. Go to www.angelmanbehaviors.org
2. Complete a short profile
3. Access each segment or module individually and explore its contents

Angelman Family Contributions: Behavior

Distraction is our best trick for preventing our daughter's screaming or arching of her back. The word help is magical "Elena please help me with_____." This must be said before the disruptive behavior begins. For example, Elena always seems to scream or arch her back when going from her room to the kitchen, or from the family room to the garage. If I say, before going down, "Please Elena could you help me carry this?" she feels useful and no disruptive behavior appears.

Maria, Madrid, Spain, angel Elena, age 35

My son's non-compliant behavior used to frustrate me greatly. Now I give him more time to respond to my requests and it has made a big difference. When I ask him to come to the table, get out of bed, get dressed etc. and he does not respond, I walk away and ask him again in a few minutes. I may have to do this a few times, but eventually he responds and does what I ask of him.

Inge ingediehl@hotmail.com, angel Marcus, age 34, Del+

Be very careful about over stimulation! Too much noise, too many people, too many toys out at once, etc. My angel gets over-stimulated with too much of just about anything. This causes behavioral issues for us.

Elise

Emotional reactions to challenging behaviors can be very reinforcing. In our experience, keeping cool and minimizing engagement – including eye contact, physical contact, and talking – during challenging episodes really helps.

AS Family Member

Using a weighted blanket has helped the child I work with relax and become calm when upset.

Ann

Our angel just gets absolutely wild sometimes – pulling things off the shelf, throwing things, and pushing and hitting. I have found that holding her really tight from behind and whispering to her or singing in a very soft slow tone will calm her. It is like pushing a reset button! It usually takes a minute or two of this to completely calm her and she is resistant at first!

Elise

Through the years, we have learned that it is best to be patient and allow a few quiet moments for our angel to respond to directions and process the information. This is especially true when using the restroom, moving from one place to another, beginning to eat breakfast, etc.

AS Family Member

ABA therapy has helped our son's behavior greatly, as well as having a Board Certified Behavior Analyst involved in all aspects of our son's care. Our kids use their behavior to communicate. Our BCBA helps us figure out what our son is trying to tell us with his behavior. Our son's behaviors have gotten more intense as he has gotten older. I have found that people who are trained and want to work with individuals who have challenging behaviors are the only ones that end up staying with us for the long haul.

AS Family Member

Let your angel know it hurts when they pull your hair. Let bystanders, friends, family know she pulls hair and dangling things. Reinforce that it's not appropriate.

Norma, Murrieta, CA, angel Katarina, age 20, Del +

Keep in mind that much of the "behaviors" are the result of anxiety, frustration, lack of sleep, illness, etc. Certainly, our angels suffer "fight or flight" at times and this can be traumatic for all. The harder, but more effective strategy, is to whisper reassurance, let them know that everything will soon be OK and that you are there. Patience is the key- even in public when things might become embarrassing. (Most people will be sympathetic.) Remember... "This too will pass." It will take a while - and longer than you want - for your child to calm down. He/she might lash out at loved ones with slapping or hitting. Do not take it personally! It is often beyond their control. As much as we try, we cannot imagine what it is like to have Angelman syndrome. NOT resorting to physical discipline or screaming (which increases aggression and frustration) will, in the long run, help you as a parent feel better about yourself as a parent and will deepen your child's trust in you. As Dr. Phil says, "Children need a safe place to land" and you can create that by using comfort rather than anger.

Alice, sandiegoasfwalk@gmail.com, San Diego, CA, angel Whitney, age 38, Del+ Class 1

Due to the fact that most children with AS do not speak you should start ABA at an early age. "ABA" is Applied Behavior Analysis. They come to your home or you can take your child to a clinic to get these services. It is a fantastic service that really will give your child a better way to communicate, interact appropriately with peers and family and so much more. It has made a huge difference in my son's life. We have been doing it for 6 years now.

Duane, angel Aiden, age 12, Del+

When my son gets really worked up and cannot be soothed, we try to change the activity or environment so that he can regulate his behavior and emotions before we go back to resolving the issue (if it needs resolved). One way that we help him to focus on something other than what has upset him, is to have him run his hands under lukewarm/cold water. Helps dry up tears quickly! We use this method a lot for boo-boos.

Desiree, desireemartika@yahoo.com, angel Titus, age 3, Del+

I have found that the unusual laughing associated with my son's Angelman syndrome is directly related to his need to use the restroom. It's almost a form of communication or nervousness. Once he has used the restroom, the laughing stops.

Nancy, Lebanon, Ohio, angel Paul, age 29

BIKES & TRIKES

TRIAID Cycles for Life <https://www.triaid.com/>

TRIAID is the manufacturer and supplier of special needs equipment for children, specializing in special needs tricycles and handcycles.

Sand Rider- Beach Wheelchairs <https://custombeachwheelchair.com/>

A new beach wheelchair to ensure that everyone can enjoy being out of doors for the day, swim at the beach or share off-road activities on gravel trails, grassy fields, swampy areas or snow.

Angelman Family Contributions: Bikes and Trikes

My son rides an adult *Rifton* trike. He loves it! It is great exercise!

AS Family Member

Helping our Angels ride a bike safely can be a real challenge. We tried a three wheel adult sized trike but our son would turn off the sidewalk and ride right into the street, even with Mom or Dad riding (or running) alongside. Scratch that. So next we tried a tandem with our son riding on the back. Balancing the bike with a wiggling and wobbling child on the back was waaaaay too dangerous! Scratch that. Then we found the Rhoades Car Quadricycle. Four wheels, low to the ground, two chair like seats with backs sitting side-by-side (with option to add two rear passenger seats which sister and the neighbor kids loved), both front passengers have pedals but only one with the handlebars to control the steering!

Finally, a safe option that allowed our family to bike together safely AND allowed our son to pedal and get much needed physical activity. His sister and her friends loved it, too! And we won the best decorated bike in the neighborhood parade 2 years in a row! More information can be found on their webpage at rholdescar.com

Elaine, MARTINEK55@YAHOO.COM, angel Paul Deceased at age 32 in 2010, Del+

We bought a standard toddler trike (the ones that have a harness and can be found at Target) for our two year-old grandson. He couldn't push the pedals, so we used a cable to tie a pair of sandals to the pedals. We strap his feet in his sandals and away we go pushing him! It has improved his leg muscles and he loves going for rides.

Dimity

[Our son] started riding an adapted tricycle until his legs got stronger. By age 6 he was riding a regular bike with training wheels that his brother passed down to him. To help [him] feel comfortably balanced on the bike with training wheels, we bought a wide adult bike seat from Wal-Mart and put it on his bike. His whole bottom end fits on the seat and he feels balanced on his bike.

AS Family Member

BODY TEMPERATURE REGULATION

Individuals with AS have poor body temperature regulation and increased sensitivity to heat.

This abstract appeared in an international 2008 PubMed article:

An 8-month-old girl and a 20-month-old boy with Angelman syndrome and long-standing fever are presented. Despite extensive clinical and laboratory examinations, no inflammatory or infectious origin for the fever was found. It was considered that the long-standing fever observed in these cases was due to *hypothalamic dysfunction for thermoregulation*.

Angelman Family Contributions: Body Temperature Regulation

Several years ago, our son David had a sore throat and would not eat or drink. This was behavior he had exhibited in earlier bouts with sore throats, so we weren't too concerned. He also didn't seem really sick and to our touch his body didn't seem too warm. To our horror, as he was sitting at the dinner table, he collapsed. We rushed him to the emergency room to find that he was running a temperature of 108 degrees. Due to the wonderful doctors and care he was given, he survived; but he ended up in the hospital for five weeks. The doctors told us that there was a problem with his "temperature regulator" and consequently he could spike a seriously high fever very quickly

Fred and Carolyn

Last summer my husband bought my daughter a personal fan/mister. We took this everywhere, so that we could still do all the things we love outside...going to the beach, going on walks, spending an afternoon at the zoo. She thought the fan was funny, so we got to hear her great laugh, as well as keep her cool.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

C

CAMPS

Center for Courageous Kids: Family Retreat for special needs families.

1501 Burnley Road Scottsville, KY 42164 info@courageouskids.org www.courageouskids.org

My Summer Camps: Resource link for finding summer camps for Special Needs.

https://www.mysummercamps.com/camps/Special_Needs_Camps/

Other Camps listed by state in ABC order:

Adaptive Aquatics Alabama <http://www.adaptiveaquatics.org/default.aspx?id=2> Adaptive Aquatics is a 501(c)(3) nonprofit organization dedicated to the introduction, teaching and advancement of adapted water skiing for children and adults with physical disabilities.

Welcome to Easter Seals Camp ASCCA Alabama <http://www.campasca.org/> Camp ASCCA is **Alabama's** Special Camp for Children and Adults. ASCCA is a nationally recognized leader in therapeutic recreation for children and adults with both physical and mental disabilities. Providing weekend and week-long sessions, Camp ASCCA is open year-round.

Camp Smile/Life Without Limits Alabama <http://www.campsmilemobile.org/> The summer camp experience is about much more than horseback riding, kayaking and swimming in the lake. For young people across America, summer camp is a stepping stone on the road towards independence. Nowhere is this truer than at Camp SMILE. Hosted by United Cerebral Palsy of Mobile, Camp SMILE is a residential summer camp for individuals (ages 5-10) with and without disabilities. Here, campers not only have the chance to participate in physical activities like kayaking, fishing, and horseback riding, they also meet others whose abilities and life experience are similar to their own.

Lakeshore Foundation Alabama <http://www.lakeshore.org/> Lakeshore Foundation is a 501(c)(3) organization that promotes independence for persons with physically disabling conditions and provides opportunities to pursue active, healthy lifestyles. Lakeshore Foundation offers a wide range of fitness, recreation, athletic and education programs to children and adults who experience diagnostic conditions including spinal cord injuries, cerebral palsy, multiple sclerosis, stroke, amputation, and visual impairment. The Foundation also serves persons who have been diagnosed with arthritis, diabetes, chronic pain, cardiac conditions, and many other related disorders.

Challenge Alaska Anchorage, AK <http://www.challengealaska.org> Challenge Alaska is based in Anchorage and offers therapeutic recreation, sports and education for people with physical and mental disabilities.

SAIL/ORCA Alaska <http://www.sailinc.org/orca.php> ORCA (Outdoor Recreation and Community Access) is the recreation program of SAIL. ORCA's purpose is to promote inclusive recreation and adaptive outdoor pursuits for Southwest Alaskans who experience disabilities. Our philosophy is that recreation influences all aspects of a person's life: their employment, social life, physical and spiritual well-being and a sense of place in their community. Challenging recreation opportunities give an individual a chance to recognize their ability to succeed.

Special Olympics Arizona <http://www.specialolympicsarizona.org/> Provides every person with intellectual disabilities a place of welcome, acceptance and the chance to be their best: Special Olympics Arizona is made up of passionate, committed individuals from every walk of life, who recognize the value and unique gifts of people with intellectual disabilities. Together, they share the common belief in dignity, equality, and opportunity for ALL people.

Arizona Disabled Sports <http://www.arizonadisabledsports.com/> The Mesa Association of Sports for the Disabled (MASD) is a nonprofit organization dedicated to providing sports and recreation opportunities to individuals with all types of disabilities. This could include but not be limited to developmental disabilities, orthopedic impairment, sensory deficit and/or neurological involvement. The Association provides year round sports training and competition through our Special Olympics programs for athletes who are intellectually disabled. In addition, sports and recreation opportunities are provided for individuals with physical disabilities through our programs sanctioned by Wheelchair & Ambulatory Sports, USA; BlazeSports America; Disabled Sports, USA; and the United States Paralympics.

Free Surf Camp for Special Needs, La Jolla, CA

<http://specialneedsresourcefoundationofsandiego.org/free-surf-camp-for-kids-with-special-needs/>
<https://www.autismsocietysandiego.org/surf-camp.html>

Foundation for Dreams, Dream Oaks Camp, Bradenton, FL <http://www.foundationfordreams.org/> The Mission of the Foundation for Dreams, Inc. and Dream Oaks Camp is to provide fun, educational and recreational experiences in an outdoor camp environment to enhance the lives of children ages 7-17 with physical and developmental disabilities and serious illnesses.

Quest's Camp Thunderbird, Apopka, Florida <https://www.veryspecialcamps.com/summer-camps/Quests-Camp-Thunderbird-2160.html> Because of the physical and behavioral challenges associated with Down syndrome, autism, cerebral palsy and other developmental disabilities, the opportunities for these individuals to relax, socialize and have fun are often very limited. Quest's Camp Thunderbird's six- and 12-day overnight sessions gives parents and guardians a break from the demands of 24-hour caregiving. For many families, this respite is their first chance to relax, spend time with friends, or even take a vacation knowing that their loved one is receiving quality care and attention.

Camp Twin Lakes, Atlanta, GA <http://www.camptwinlakes.org/> Camp Twin Lakes is a unique non-profit organization in Georgia with campsites, day camps and hospital-based camp programs designed specifically for children with serious illnesses, disabilities and other challenges. In collaboration with its network of Camp Partners, Camp Twin Lakes provides an enriching camping experience for children who might otherwise be unable to attend camp. CTL was founded in 1991 and began camp operations in 1993 with seven camp groups. Since 1993, Camp Twin Lakes has served well over 40,000 campers.

Camp Caglewood, Flowery Branch, Georgia <https://www.veryspecialcamps.com/summer-camps/Camp-Caglewood-2207.html> Caglewood is a special needs camping program that serves

individuals with developmental disabilities through active programming that provides enhanced life experiences, personal development, and respite opportunities. Caglewood fosters the core development of those they serve by engaging them in activities designed to promote independence and confidence. Caglewood's programming inspires spiritual and social development through communion with nature. A camping trip with Camp Caglewood consists of counselor-planned and led activities, crafts, devotionals, hiking, camping, swimming, and various forms of physical therapy and adaptive skills training. Caglewood is designed for both children and adults diagnosed with developmental disabilities including the autism spectrum, PDD, Down syndrome, and cerebral palsy.

Camp Imua, Wailuku, HI <https://imuafamilyservices.org> A Special Camp for Special Campers, Camp Imua is an incredible Maui tradition that began in 1976. Held each year in June at Camp Maluhia in the West Maui Mountains, fifty campers and over one hundred volunteers come together for a week of crafts, games, swimming, helicopter rides and general fun, all the while making new friends and catching up with old. Camp Imua brings the community and children with special needs together for a free weeklong, overnight recreational camp each summer. The Camp gives school-age children with "special abilities" opportunities to try new activities and connect with others. It gives volunteers opportunities to connect with the children and to give back to the community. Camp Imua also gives the children's caregivers much needed time off. Fifty children attend and at least 150 community volunteers and numerous businesses from restaurants, helicopter companies, and live bands make Camp Imua memorable and fun for all. Over 70% of Camp's expenses are met through in-kind donations.

Camp Callahan, Inc., Lima, IL <http://www.campcallahan.com/> Through the agreement with the Boy Scout Council, the Camp Callahan Program is conducted at Saukenauk Scout Reservation located 25 miles north of Quincy and 4 miles east of Lima, Illinois. It is the property of the Mississippi Valley Council. Saukenauk is 602 acres of prime Boy Scout Real Estate that includes four camps. Camp Callahan, Incorporated, now in its 54th year of service to people with disabilities, offers a program that provides an exciting, safe, summer camp experience. Camp Callahan Inc., their volunteers, and the facilities of the Mississippi Valley Council of Boy Scouts of America, will combine to make this unique program a success. Camp Callahan is dedicated to serving youth with wide-ranging disabilities through a stimulating, liberating program in an environment that is physically and socially adapted to the personal needs of these very special campers. The camp has the specialized environment, equipment, activities, accessible buildings, and a trained adult staff to overcome the barriers that a person with disabilities would encounter at ordinary summer camps.

Anderson Woods, Inc., Bristow, IN <http://www.andersonwoods.org/> Anderson Woods provides summer camp experience and safe, dignified, permanent residences for persons with mental and/or physical disabilities. They strive to teach residents (to the maximum extent possible) to be fully functioning members of the local communities. Since 1979 Anderson Woods has been hosting those with special needs during the summer. Campers learn self-confidence, trust and responsibility through working together, caring for animals, tending the garden, and enjoying the beauty of nature. Camp is a place where everyone is equal. We embrace all races and spiritual beliefs.

Camp Discovery, TN <http://www.jayceecamp.org/> The Tennessee Jaycees Foundation, Inc. was incorporated on December 11, 1972, and was officially organized by the incorporators on February 17, 1977. It has been classified as a charitable organization by the Internal Revenue Service, meeting the requirements of Section 501(c)(3) of the IRS Code. The IRS has determined that the organization is not a private foundation (in other words, it receives its support from the general public). Camp Discovery is a camp for mentally and physically challenged kids and adults to experience the wonders of the summer

camp setting. If you know of a person with special needs interested in having a life affirming experience at our camp, please direct them to our application posted on the website.

Vanderbilt Kennedy Center, Tennessee <https://vkc.mc.vanderbilt.edu/vkc/> The Vanderbilt Kennedy Center offers a variety of summer programs for children, adolescents, and young adults with and without disabilities. Our camps are a wonderful way to encourage self-esteem, self-respect, and compassion while learning valuable life skills, making new friends and discovering new interests. Vanderbilt Kennedy Center programs are unique in that they provide model services for participants, support for families, training opportunities for college students preparing for educational or service careers, and opportunities for participant and family members to take part in innovative research.

Morgan's Wonderland, San Antonio, TX <http://www.morganswonderland.com/index.html> Morgan's Wonderland was built in the true spirit of inclusion to provide a place where all ages and abilities can come together and play in a fun and safe environment. Morgan's Wonderland, the world's first ultra-accessible family fun park, encompasses 25 acres of rides, attractions and activities for everyone, and all are welcome! Come play with us!

Camp Koinonia, Virginia Tech, <http://www.thecampkoinnia.com/> Camp Koinonia is an outdoor education program for children ages 7-22 who have multiple disabilities. The program was developed in 1977 at Virginia Tech as part of a class with the primary purpose of providing a meaningful, experiential learning opportunity for university students while involving children and young adults, some with severe disabilities, in outdoor activities that they would not be able to do otherwise. During the first year of the program a counselor/camper duo submitted the winning name for the program – Camp Koinonia. Koinonia comes from the Greek and means 'fellowship' and 'caring community'. Since that time the purpose and mission of Camp Koinonia has been to provide a 'caring community' for their campers in sense of true 'fellowship'.

Angelman Family Contribution: Camps

It can be scary to let them be at camp but our son greatly enjoyed it, once he understood what it was after the first day, at camp. Our local university offered one with one to one counselors and we could often use respite funds to cover the expense and ask for scholarships through the program. The social aspect is great and they can work on communication, physical and independent living skills. It's an amazing break for parents and siblings! Nursing staff are on hand for needs.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18 Del+ Class 1

CAUSES OF ANGELMAN SYNDROME

Deletions 15q11.2-q13 (68% of cases) – the majority of AS cases are caused by deletions on the maternal copy of Chromosome 15. Due to genomic imprinting, only the maternal copy of UBE3A is expressed in the brain. The deletion thus removes the normal expression of this gene in AS individuals. Class 1 refers to individuals with larger deletions. Class 2 is smaller deletions. *See D- Deletion Positive*

UBE3A mutations (11% of cases) – In these individuals, mutations in the UBE3A gene either prevent its expression or function. Thus these individuals do not have the appropriate levels of functional UBE3A in the brain. *See U- UBE3A*

Uniparental disomy (UPD; 7% of cases) – in UPD, the individual has two copies of paternal Chromosome

15. Because UBE3A is not expressed from the paternal copy, these individuals lack normal levels of UBE3A in the brain. *See U-UPD*

Imprinting defect (3% of cases) – These individuals may have a deletion of the imprinting center on Chromosome 15, but cases can also be caused by loss of imprinting information during the mother’s oogenesis. Loss of imprinting will prevent expression of the maternal UBE3A gene in the brain. *See I-Imprinting Defect*

Clinical/other (11%) – In these individuals, all testing for Angelman syndrome is normal, but they still meet the diagnostic criteria for AS. These individuals may have as yet unrecognized mutations.

Recurrence Risks

UBE3A or genomic imprinting on Chromosome 15 - An increased risk seems likely but probably does not exceed 10%.

Germ Cell Mosaicism - This term refers to a phenomenon in which a genetic defect is present in the cells of the gonad (ovary in the mother’s case) but not in other cells of the body. This occurrence can lead to errors in risk assessment because a genetic test, for example on a mother’s blood cells, will be normal when in fact a genetic defect is present in the germline cells of her ovary. Fortunately, germ cell mosaicism occurs very infrequently. Nevertheless, it has been observed in AS caused by the mechanisms of large chromosome deletion, Imprinting Center deletion and UBE3A mutation. *See G-Germ Cell Mosaicism*

Imprinting Inheritance - UBE3A mutations and Imprinting Center deletions can exhibit imprinting inheritance wherein a carrier father can pass on the genetic defect to his children without it causing any problems, but whenever a female passes this same genetic defect on to her children, regardless of the sex of her child, that child will have AS. The pedigree diagram below illustrates imprinting inheritance. Here, AS has only occurred after a carrier mother passed on the gene defect (for example as in the two siblings with AS pictured on the left lower part of the pedigree). In addition, a distant cousin in this family also has AS due to the imprinting inheritance. When an AS genetic mechanism is determined to be inherited, genetic testing of family members can usually identify carriers of the gene defect. As you might imagine, professional genetic counseling is advised in these situations.

See I- Imprinting Inheritance

***Also, see this excellent link from the University of Florida:

https://www.peds.ufl.edu/divisions/genetics/programs/angelman_syndrome/genetic_counseling.htm

It details recurrence risks:

- Common Chromosome Deletion less than 1% recurrence risk
- Paternal Uniparental Disomy less than 1% recurrence risk
- Imprinting Center 50% recurrence risk
- UBE3A Mutations 50% recurrence risk
- Individuals with no known mechanism (does not exceed 10% risk)
- Germ Cell Mosaicism
- Imprinting Inheritance

CBD OIL

CBD May Alleviate Seizures, Benefit Behaviors in People with Neurodevelopmental Conditions

<https://angelman.org/cbd-may-alleviate-seizures/>

Patients and families should always seek advice from their physician before taking any CBD products, and know that a human clinical trial is needed to fully understand its efficacy and safety.

Angelman Family Contribution: CBD Oil

Charlotte's Web worked for us. Wings did not given its THC content, but we have heard great success stories from other families so it really is trial and error for each individual body and see how it responds.
Tatiana, angel Alina, age 4, UBE3A Mutation

CELEBRATIONS

Angelman Family Contributions: Celebrations

We celebrate all positive behavior, efforts, and ALL milestones. We focus on encouraging through positive affirmations, singing, clapping, dance parties, "woohoo's!" We have found this to be a huge self-esteem boost, & motivation for all of the growth & development.

Annie, Folsom, CA, Ava, 7 Del+

Bouncy houses have been a great hit with our Angel, it also creates an activity where she is included with other children.

Tatiana, angel Alina, age 4, UBE3A Mutation

When young, our son did not enjoy loud singing or yelling. We would softly sing *Happy Birthday* or warn him in restaurants, etc. Silly distractions can help. We also used a lot of praise for skills gained and small triumphs as they seem to love positive feedback!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18 Del+ Class 1

CHAMPIONS - ASF

See the list of Champions on our website. Champions are AS parents who are serving as ASF "Champions". Please contact one in your area for specific information and support. They are eager to help!

CLINICS

See A- ASF Clinics

CLOTHING

Angelman Family Contributions: Clothing

For my daughter, clothing is a real challenge. She seems to feel very uncomfortable in different fabrics and even different styles of shirts. Here's my tip: consignment stores! I will go often, especially during sales, and buy a variety of tops and dresses that I think my daughter will like. My daughter pretty quickly shows me what she doesn't like by pulling at the collar, fussing, or chewing on the item. By utilizing consignment stores, I can find all kinds of tops in different brands and styles. I will often

discover a favorite item, and then I look up that style and brand online and purchase duplicates from places Poshmark or even the original retailer, like Target.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del +

Dress them with age appropriate clothes.

Norma, angel Katarina, age 20, Del +

For night time... I cut open the back of a t-shirt and sew in a zipper. Then I sew the t-shirt to elasticized shorts or long pants. It is now a onesie that can actually be worn in public places. You could use an AS shirt to spread awareness.

Cheryl, weitty6@yahoo.com, angel Megan, age 23, Del+

An Australian company, named Wonsies, make great one piece pajamas for an age, size, or weight and some designs are escape-proof! wonsie.com.au

Pam and Warren, wpnew@windstream.net, angels Jonathan and Andrea, ages 40 and 35, UBE3A Mutation

The easier the clothing the better. Choose pants without zippers and buttons. He wears an adult one-piece footless sleeper that we put on backwards to prevent diaper digging at night.

AS Family Member

I recommend "Life is Good" t-shirts. They are sturdy around the neck and are well made. An added bonus is they can feature a written inspirational message which makes your child an "ambassador" and it gives them a "voice". Part of the proceeds go to the *Life is Good Foundation* which helps children in need. P.S. *The ASF t-shirts are great, too!*

Alice, sandiegoasfwalk@gmail.com, San Diego, CA, angel Whitney, age 38 Del+ Class 1

COMMUNICATION TRAINING SERIES

<https://www.angelman.org/resources-education/communication-training-series/>

The Communication Training Series was developed to break down often very complex communication training and instruction into smaller, simpler parts, where care providers and their support teams can go at their own pace as they are working with their special person with AS. The series includes training tools and how to use picture symbols, includes downloadable picture and symbol display templates and step-by-step instructions to start and continue a communication program at home and school, no matter the individual's age, skill level or location. The ASF Communication Series was developed by a leading team of experienced communication experts in AS and other non-verbal populations.

To date the ASF educational webinar and communication training series have collectively seen over 75,000 views. The series continue to be used extensively throughout the global AS community and a future goal is to translate the ASF Communication Series into Spanish.

Book

So Much to Say by Dr. Stephen Calculator

Dr. Calculator goes beyond step-by-step instructions – he gets to the root of why, how, when and where. His philosophy is built upon presuming confidence and the fundamental belief that overestimating potential has less negative consequences than underestimating it.

So Much to Say arms you with a lifetime of experiences that have created more effective communicators who are better able to participate in all facets of life. And as a parent, you know what that means: more happiness, more confidence and more self-ability for your loved one.

Details: Published April 3, 2018 Paperback – 308 pages \$18.99

To purchase: <https://www.angelman.org/resources-education/communication-book/>

Communication Strategies for Children with Angelman syndrome

Augmentative and alternative communication (AAC)

<https://consultqd.clevelandclinic.org/communication-strategies-for-children-with-angelman-syndrome/>

Authors: Ms. Kearns is a senior speech-language pathologist and Coordinator of the Technology Resource Center in Cleveland Clinic Children's Hospital for Rehabilitation. She works with children with a variety of complex communication needs. Dr. Henry is Director of Developmental Pediatrics and Physical Medicine and Rehabilitation.

Angelman Family Contributions: Communication

This is the aspect of AS that causes me the most sadness even though our son has over sixty signs. He shakes his head yes and no and uses an AAC device. Nothing can ever replace verbal speech. If he could just tell me "Mom, I do not feel well... Mom, I do not like that person... or, Mom, I am soooo bored at school," that would make his life so much easier.

AS Family Member

Let your angel know what is coming up next. *When you are done eating we are going to brush your teeth." Give them time to process. "When I'm done with my coffee, you are going to get a bath."

Norma, Murrieta, CA, angel Katarina, age 20, Del +

Our son has done very well learning the LAMP AAC program for the iPad. It is a motor-planning concept that works well for kids with Autism. He has learned where words are located and only has to push 1-2 icons to make a word appear in the text line and hear it voiced. We have enjoyed knowing what he's interested in and being able to verbally interact, and he often likes to make funny comments!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18 Del+ Class 1

Communicate by talking simply, directly, and use visuals as much as possible while talking even if your Angel is nonverbal. This helps with lowering anxiety and his/her uncertainty. Assume he/she might respond even when you know he won't.

Terry, angel Byron, age 35

My daughter's communication is very limited... no words, no pointing. My tip is to focus on what our kids can communicate and build on that. When she smiles, I smile back and hug her. I assume she's telling me she loves me. When she laughs at something during dinner, we all pause and reflect back to her and why she must be laughing out, usually causing us all to laugh more at the many theories of what it might be. When she sneaks past the den and ends up feasting on bananas on the kitchen counter, I don't get mad (though this ruins her keto diet), I laugh and let her know that I know she must miss this favorite food and I try to add a little to her next meal. When she fusses, I try to figure it out and if I solve the puzzle of her frustration, she usually smiles up at me, as if to say, "Thank you. You heard me"

Sarah, bnamommysifun@yahoo.com, angel Lily, age 14, Del+

Throughout my daughter's life, I have used a special voice when talking to her. Her eyes always light up with a big smile because she clearly knows I am having a conversation with HER! I talk to her as if she understands every word. I also admit that I talk FOR her at times. This adds humor to our daily lives, and I strongly believe it empowers her when I read her mind!

Alice, sandiegoasfwalk@gmail.com, San Diego, CA, angel Whitney, age 38, Del+ Class 1

At the beginning of the day, I narrate to my son what the day will be like just as you would tell a typical child the big events on the schedule (Ex. "We're headed to daycare, and after breakfast Mommy will come and get you for therapy. After therapy, you go back to daycare for lunch and a nap. At the end of the day, Daddy will pick you up and we'll take you to ride the horse." After each event, we, again, narrate the rest of the day to remind him. Time and time again, he has proven that he understands (3 yr old, del+) because when the schedule changes and someone doesn't show up after breakfast, he throws a fit and keeps crawling toward the gate where parents arrive.

Desiree, desireemartika@yahoo.com, angel Titus, age 3, Del+

COMMUNICATION ADVISORY COMMITTEE (CAC)

<https://www.angelman.org/about/communication-advisory-committee/>

The Angelman Syndrome Foundation has developed a Communication Advisory Committee (CAC) committed to helping all individuals with Angelman syndrome improve their communication skills. The CAC is made up of eight Angelman syndrome, education and communication industry experts. The team works toward two major goals: to define high-priority areas of AS communication research for funding, and attract new professionals to explore AS communication research. The CAC will develop a strategic plan for use by the ASF Board of Directors and Scientific Advisory Committee to guide the review and funding of communication research proposals.

Dan Harvey, PhD CAC Chair

Dan Harvey was appointed to the ASF Board of Directors in 2012, and chairs the ASF Scientific Advisory Committee and Communication Advisory Committee. Dan has more than 20 years of experience in drug discovery research both in academia and the pharmaceutical industry. He became involved with the ASF after his son, Matthew, was diagnosed with Angelman syndrome in 1996. From 1997 to 2001, Dan was a member on the ASF Board of Directors and served as vice president from 1997 to 1999. Dan resides in San Diego, California with his wife, Karen, and has three children, Michelle, Jay and Matthew. Matthew is diagnosed with AS.

Stephen Calculator, PhD

Stephen Calculator was appointed to the ASF Scientific Advisory Committee in 2008 and most recently selected as a member of the Communications Advisory Committee.

Jane Summers, PhD

Jane was appointed to the ASF Scientific Advisory Committee in 2006 and will bring her expertise to the Communications Advisory Committee.

Erin Sheldon, MEd

Erin conducts professional development workshops and webinars for educators and therapists on the assistive technology, communication, and literacy needs of students with significant disabilities,

including AS. She has published journal articles, book chapters, and manuals for educators. Erin is the mother of Maggie, who is deletion positive.

Penelope Hatch, PhD

Penny Hatch is a Research Assistant Professor for the Center for Literacy and Disability Studies (CLDS) at the University of North Carolina (UNC) Chapel Hill, School of Medicine. Dr. Hatch received her Ph.D. in Speech and Hearing Sciences from the University of North Carolina at Chapel Hill.

Carole Zangari, PhD, CCC-SLP

Carole Zangari is a Speech Language Pathology (SLP) faculty member in the College of Health Care Sciences at Nova Southeastern University (NSU). Dr. Zangari's primary areas of interest are augmentative and alternative communication (AAC) for children and adults with developmental disabilities, and the use of online instruction and social media for pre-service SLPs and practicing professionals. She teaches AAC courses at the Master's and Doctoral levels, coordinates the AAC Lab, and provides clinical supervision to graduate student clinicians.

Rose Sevcik, PhD

Rose Sevcik is a Professor of Psychology at Georgia State University where she specializes in communication, language, and reading development; developmental and learning disabilities; and parent and school intervention.

Courtney Castelli, EdS

Courtney Castelli is a dedicated mom, wife and educator. Courtney has spent eighteen years in the field of education both as a classroom teacher and as building administrator. Courtney earned her Master of Science in Education from the University of Missouri – St. Louis and is in the final stages of completing her doctorate degree from McKendree University in the area of curriculum and instruction. Courtney and her husband, Eric are the parents of Lucy (AS, del +), Lauren, and Colin.

COMMUNITY

Developing a community around your child is vital. Get involved at your church and other community groups.

Angelman Family Contributions: Community

We are very actively involved in our community. Our angel Maddy, age 12, loves going to the soccer field to watch her sister play, attending professional soccer games, and going to our school's plays, musical theater productions, or dance concerts. The more we take her out into the community, the more people get to meet her and are always happy to see her smile and having a great time wherever she goes.

Myriah, Angel Madeline, age 12, Mutation

My angel Miata is 24 years old and very active in her community. We attend church, volunteer with local foundations, and are always on the go. Every weekend we are out and about, soaking up the sun and spreading joy and love to everyone we meet. We've even spoken with general managers at our local Walmart and was able to get them to order the Caroline Cart for easier shopping.

Patricia, ladyp729@yahoo.com, angel Miata, age 24

Any time we can, we include our daughter in our community events. She attends church, her brother's XC and track meets, and VBS. What we have discovered is that our daughter loves to go places, so she often has great behavior and lights up with the opportunity to be out and about with her family. On the other hand, some events are just not a blessing to her and end up being a ton of work and for something she could care less about. This is how we weed out her inclusion into community events. If I suspect she cares and wants to be a part, we make that our aim. Otherwise, we figure out a way for her to do something different or more on par with something she would enjoy. For instance, we used to have over 100 students at our house for special events. These nights simply overwhelmed her, so once we discovered this we tried to arrange something different for her, like going with a caregiver to a local store or going to our park. Another example is that for many years going to church was a huge struggle but a high priority for our family. When the time was right and leadership at our church was favorable to the idea, I started a ministry to families with special needs. This has been such a blessing because not only does it help my daughter, it expands to allow any family experiencing special needs to attend our church.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

CONFERENCES

<https://www.angelman.org/conferences-symposia/>

For information about the 2019 ASF Conference held in Louisville, KY:

<https://www.angelman.org/conferences-symposia/2019-asf-conference/>

Family Conferences: Every other year the ASF holds a conference to gather families, care providers, therapists, teachers, scientists, researchers and doctors under one roof to learn and discuss the latest information on Angelman syndrome. The conference is a great networking opportunity and a time to meet new friends and catch up with old ones! The first ASF family conference was held in Orlando, Florida in 1993.

Conferences are helpful to network with other families. Check with local agencies to see if they will fund your travel expenses. The Kiwanis Club, the Lions Club, the Optimist Club, etc. We are not too proud to ask for help with expenses...because this is a lifetime commitment. So we try to save as much of our personal money as possible for his future.

The Scientific Symposium is held every year. This two-day symposium is a chance for leading researchers, scientists and doctors to discuss the latest research activities in the world of AS. The first day is focused on a specific topic with 5-8 presenters. This day is a unique opportunity for researchers to present their latest findings of their works and compare notes with colleagues from around the globe. The second day includes short presentations on all areas of AS.

The following is a list of the ASF Family Biennial Conferences held since 1991. Also included, are the years the Scientific Symposiums were held. The Symposiums are held concurrently with the Family Conferences. ***Since 2008, the Scientific Symposiums have been held every year.

1991	Orlando
1993	Orlando
1995	Colorado Springs
1997	Seattle

- 1999 Philadelphia (Cherry Hill, NJ)
- 2001 Chicago (Oakbrook)
- 2003 Washington, DC
- 2005 Anaheim
- 2007 St. Louis
- 2008 Scientific Symposium only - Boston
- 2009 Orlando
- 2010 Scientific Symposium only – Chapel Hill, NC
- 2011 Salt Lake City
- 2012 Scientific Symposium only – Rockville, MD
- 2013 Orlando
- 2014 Scientific Symposium only – Cambridge, MA
- 2015 Chicago (Schaumburg)
- 2016 Scientific Symposium only - Silver Springs/Bethesda, MD
- 2017 Phoenix
- 2018 Scientific Symposium only – Chapel Hill, NC
- 2019 Louisville

CONSERVATORSHIP

Here is the online statement about conservatorship:

In the United States, all adults are considered capable of handling their own affairs unless a Judge determines otherwise. In California, this legal arrangement is called a conservatorship.

Conservatorships are established for impaired adults, most often older people. Adults who are developmentally disabled or the victims of a catastrophic illness or accident also may have conservatorship.

Angelman Family Contributions: Conservatorship

We went through a legal process in California to obtain conservatorship of our adult daughter in California. Our regional center case manager assisted us with the process and we appeared before a judge in Superior Court. Our adult daughter has needed several surgeries, and we have been required to bring proof of conservatorship for the surgeries to proceed. In addition, it is essential to have conservatorship with financial and other decision-making.

AS Family Member

We looked into this about 6 months before our son turned 18 to find a family lawyer and begin the process as it takes a few months but needs to be done close to the 18th birthday. The more information you can supply regarding finances and proof of AS, the smoother it will go.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

CONSTIPATION

17 Best Foods for Constipation <https://www.healthline.com/nutrition/best-foods-for-constipation>

Angelman Family Contributions: Constipation

For constipation issues, Miralax worked wonders for us. We went from once a day to every other day to twice a week. When our Angels are constipated, it can lead to fevers. This is a very gentle remedy and really does the trick.

Jeanne Seltzer

For constipation, I have found that Chia seeds work well. Use 1 part Chia seeds to 3 parts liquid (I use skim milk). Provide time to soak in a custard/pudding that can be flavored with yogurt and fruits. You could also use it when making chicken broth and vegetables.

AS Family Member

Vitafusion Fiberwell Gummies Can be purchased at pharmacies, as well as Walmart, Target, etc.). This chewable supplement provides 5 grams of fiber; has a peach, strawberry and blackberry flavor; and can be cut in half for easier chewing.

AS Family Member

This is a huge problem, especially in the early years. Our angel's extreme constipation caused complete bowel blockage which was evidenced by throwing up green bile. But eliminating dairy and having him get at least 40-50oz of water daily is a must!! Miralax every day is necessary for us.

AS Family Member

We make an effort to prevent and alleviate our daughter's constipation naturally (she is 3y 1m, del+). We try to do oatmeal several mornings a week with flax seed and usually give her sweet potatoes 3-4 times a week. Push, push, push water and other fluids. Juices never worked for her. Figs worked too well. She loves dairy and bananas, so keeping her regular has already proven difficult. When she starts having difficulty we start small with a sprinkle of Miralax in her morning juice and if we're too late we end up having to give Pedilax (suppository). She has low muscle tone in her lower abdomen and lower back. I assume that is the reason that her constipation results in a buildup at the base of her lower intestine. The suppository generally lets it slide out versus breaking it down. I know that the older she gets, the harder it is going to be to control for her, but that is why we're trying to figure out what works best now. We will eventually be trying Senna tea when she is older, as long as it doesn't interact with anything else and Probiotic supplements. We are also trying to avoid giving her sugary drinks since she will want to replace her regular drinks with sugary one once she discovers them! Best of luck!

Candace, angel Everly, age 3, Del+

CONTACT INFORMATION FOR THE ASF

<https://www.angelman.org/about/contact-asf/>

Email: Complete form at this link or email info@angelman.org

Angelman Syndrome Foundation
75 Executive Drive, Suite 327
Aurora, IL 60504

Phone: (630) 978-4245 | 800-432-6435

Fax: (630) 978-7408

ASF Family Resource Team: Complete the form at this link to email one of the ASF Family Resource team members. <https://www.angelman.org/resources-education/asf-family-resource-team/>

COPING

See H-Humor

See P- Parents' Health

See Q-Quotes

10 Tips for Parenting Children with Special Needs

Jessica Graham

Be a “professional” patient. Show up to appointments on time. Bring the necessary records. Call when you’re running late or need to cancel.

Be kind. Be friendly and polite to the medical staff. Make friends with them, not because you’re trying to manipulate them, but because you need friends.

Be an expert. Know everything you can about your child’s condition. Read journals. Study websites. Learn. You aren’t going to have time for any of this — do it anyway.

Be your child’s best advocate. Don’t be afraid to speak up. To ask questions. To get multiple opinions. To ask your doctor about the research. Take your child to the best. Switch doctors when you don’t like the standard of care.

Let the doctor be the doctor. Do this not because this person has the medical degree, but because it’s his or her job, not yours.

Don't let your child's needs isolate you. Having a child with special needs can be lonely and intimidating. Do not allow it to alienate you from the people around you.

Find support groups. Join online groups. Find a support group. Have someone to whom you can vent and relate. Don’t lose the people around you.

Allow yourself to recuperate. Whenever athletes do any kind of strenuous exercise, they rest to give their bodies a chance to recuperate and to avoid injury. Allow yourself to do the same — mental exhaustion is real.

Appreciate that things improve with time. Your child’s prognosis may not improve, his or her condition may be debilitating, and these are hard, hard things. But some things do get easier with time. Time, for better or worse, means more experience, more practice, greater perspective.

Learn how to be a parent, not just a caregiver. You’re a pharmaceutical dispensary, a home therapist, an insurance specialist (yes, you will spend so much time on the phone with the insurance company that your ears will bleed) and medical transporter. But you’re also a parent. Take the time to appreciate your child and to love that child as only a parent can.

Angelman Family Contributions: Coping

Get a good talk therapist for yourself and go regularly.

AS Family Member

CORTICAL VISUAL IMPAIRMENT

Cortical visual impairment (CVI) is a form of visual impairment that is caused by a brain problem rather than an eye problem.

The ASF has awarded a grant to Dr. Karen Erickson (2019):

The Prevalence and Form of CVI in Angelman syndrome, Karen Erickson, PhD, UNC Chapel Hill, North Carolina

Summary of Dr. Erickson’s Study

There is good reason to believe that many individuals with AS have cortical visual impairment (CVI). This is a specific type of vision impairment that may affect balance, walking, communication and behavior. This study, will accomplish three things:

- Estimate the number of individuals with AS that have CVI
- Assess the severity of CVI in individuals with AS
- Determine whether CVI has an effect on communication in children with AS

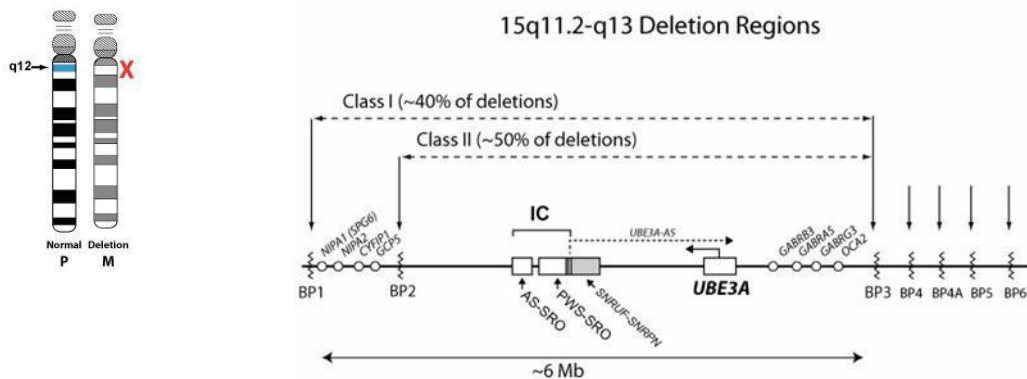
Dr. Erickson believes that in identifying and understanding CVI in individuals with AS, it will allow for early intervention, which may improve communication and behavior outcomes.

D

DELETION POSITIVE

Deletion 15q11.2-q13 (68% of cases) – The majority of AS cases are caused by deletions on the maternal copy of Chromosome 15. Due to genomic imprinting, typically only the maternal copy of UBE3A is expressed in the brain. The deletion thus removes the normal expression of this gene in AS individuals. The larger deletions has been termed the Class I deletion, and the smaller has been called Class II deletions. Class III deletions are those that are atypical and often are larger than even the class I deletions. There is less than 1% recurrence risk

<http://www.ncbi.nlm.nih.gov/books/NBK1144/> Aditi I Dagli, MD, Jennifer Mueller, MS, CGC, and Charles A Williams, MD.



Angelman Family Contribution: Deletion Positive

My 2 year old son is deletion positive! Brantley has his own strengths! He's his own individual. Don't dwell on everything online tells us. These kids can amaze us with all the challenges and hurdles they face! Just take it one day at a time!

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

DENTAL WORK

Angelman Family Contributions: Dental Work

At age 32, our UPD angel needed to have her wisdom teeth removed. This was done as an outpatient surgical procedure at the hospital while under general anesthesia. It was helpful that she was given anti-anxiety medication by the anesthesiologist while in pre-op. The oral surgeon used extra self-absorbing stitches and “glue”. (I was so concerned about her putting her fingers in her mouth post-surgery, or going into gag mode but that didn’t happen). Due to her diagnosis, I was allowed to be in the post-op room as she was waking up and followed her into the recovery room as well. When home, she slept A LOT and ate very little. Jell-O went down okay. It took a day or two to tolerate mashed potatoes. Not to use a straw to drink. The prescribed pain medication tablets were too hard for her to swallow and she did better with Tylenol ES in gel caps form. We used bags of frozen peas or baggies of ice in winter weight socks to place on her jaw (which she surprisingly enjoyed). She experienced swelling of her lips which we were told was a normal post-surgical possibility. And she did bite her lips while they were numb which didn’t help the situation. There was one prolonged episode of vomiting as well from swallowing blood. Carmex lip balm was soothing. Overall, she did well. It took 7-10 days until she was back to herself again. I think it was also helpful that she likes watching medical things on YouTube and was viewing shows about what happens at the hospital.

Kathy, angel Julienne, age 33, UPD

I think it is extremely important that you start young (around 2) with a good pediatric dentist. Our insurance covered check up every six months and we paid for extra ones to make a total of 4 visits per year. By going frequently you can establish a relationship with the provider and staff. At 37 my daughter still goes to the office she started with at 2 though the staff and dentist have changed. She knows the place and the routine and they know her.

AS Family Member

At the dental office we help the dentist along with two other people in order to do the work.

Brisia, angel Mario

Our daughter had her wisdom teeth removed at age 20. General anesthesia was used. The surgeon used extra stitches to prevent bleeding, and he also used a local anesthesia to numb the area in an effort to improve the immediate post-surgery period of time. An added bonus of the numbness was we heard her say, “Ma Ma”! :) The surgeon recommended one dose of Tylenol and one dose of Ibuprofen for pain. The process went much better than we expected! ***Also, at age 38, she is still seeing the same pediatric dentist every six months for cleaning. A dental papoose board is used. She doesn’t love it, but she is all smiles afterwards when we clap and cheer!

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del +

DIAGNOSIS: For Parent/Care Providers of Newly Diagnosed Individuals

A blood test can detect up to 80-85% of individuals with Angelman syndrome by identifying whether the UBE3A gene is functioning properly.

For the remaining 15-20% of individuals, an experienced clinician who is familiar with Angelman syndrome can provide a clinical diagnosis. Use this link to Gene Reviews to read the following information and more! It includes testing protocols.

<http://www.ncbi.nlm.nih.gov/books/NBK1144/>

Families sometimes don't know where to turn once they've received a diagnosis. It is life-changing and can be overwhelming. The ASF has been supporting Angelman families for over 25 years. We and our network of families are here for you.

Start with these easy steps:

- Fill out the form (link below). We will add you to our AS Families email list and send you a packet about AS and information about ways the ASF can help as you begin this journey.
- Use this website to become acquainted with Angelman syndrome and some of our services, like the Family Resource Team. Learn about the latest research to find treatments and a cure for Angelman syndrome.
- Connect with the world-wide Angelman community of families and supporters through our social media channels.
- Have a specific question and want to contact ASF right away? Call us at 800-432-6435 or email us at info@angelman.org.

To Complete the Form, go to: <https://www.angelman.org/what-is-as/newly-diagnosed/>

Facebook <https://www.facebook.com/AngelmanSyndromeFoundation>

Twitter <https://twitter.com/angelman>

YouTube <https://www.youtube.com/user/AngelmanSyndromeFdn>

LinkedIn <https://www.linkedin.com/company/angelman-syndrome-foundation>

Diagnostic Criteria for Angelman syndrome

Go to the Angelman Syndrome Foundation website to watch a video to learn symptoms from families of individuals with Angelman syndrome.

<https://www.angelman.org/what-is-as/testing-and-diagnosis/>

Angelman syndrome: Consensus for Diagnostic Criteria

The information and tables included below are part of a consensus paper on AS that was published in 2006 (American Journal of Medical Genetics 140A:413–418) and was the result of a work group convened by the Scientific Advisory Committee of the Angelman Syndrome Foundation. Table 1 summarizes the developmental history and Table II summarizes the main clinical features. There are no precise features that allow the physician to definitively make the diagnosis but there are characteristic features that occur with high probability in the syndrome.

Table 1. 2005: Developmental History and Laboratory Findings in AS*

- Normal prenatal and birth history with normal head circumference and absence of major birth defects. Feeding difficulties may be present in the neonate and infant.
- Developmental delay evident by 6-12 months of age, sometimes associated with truncal hypotonus. Unsteady limb movements and/or increased smiling may be evident.
- Delayed but forward progression of development (no loss of skills).
- Normal metabolic, hematologic and chemical laboratory profiles.
- Structurally normal brain using MRI or CT (may have mild cortical atrophy or dysmyelination).

* These findings are useful as inclusion criteria but deviations should not exclude diagnosis.

Table II. 2005: Clinical Features of AS

A. Consistent (100%)

- Developmental delay, functionally severe.
- 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 motion.
- Behavioral uniqueness: any combination of frequent laughter/smiling; apparent happy demeanor; easily excitable personality, often with uplifted hand-flapping or waving movements; hypermotoric behavior.
- Speech impairment, none or minimal use of words; receptive and non-verbal communication skills higher than verbal ones.

B. Frequent (nearly 80%)

- Delayed, disproportionate growth in head circumference, usually resulting in microcephaly (≤ 2 S.D. of normal OFC) by age 2 years. Microcephaly is more pronounced in those with 15q11.2-q13 deletions.
- Seizures, onset usually < 3 yrs. of age. Seizure severity usually decreases with age but the seizure disorder lasts throughout adulthood.
- Abnormal EEG, with a characteristic pattern, as mentioned in the text. The EEG abnormalities can occur in the first 2 years of life and can precede clinical features, and are often not correlated to clinical seizure events.

C. Associated (20-80%)

- Flat occiput
- Occipital groove
- Protruding tongue
- Tongue thrusting; suck/swallowing disorders
- Feeding problems and/or truncal hypotonia during infancy
- Prognathia
- Wide mouth, wide-spaced teeth
- Frequent drooling
- Excessive chewing/mouthing behaviors
- Strabismus
- Hypopigmented skin, light hair and eye color compared to family, seen only in deletion cases
- Hyperactive lower extremity deep tendon reflexes
- Uplifted, flexed arm position especially during ambulation
- Wide-based gait with pronated or valgus-positioned 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)
- Scoliosis
- Constipation

Reviewed 10-8-2015

Charlie Williams, MD

DIAPERS

SOSecure Containment Swim Brief

Made by Discovery Trekking Outfitters. An effective and discreet unisex swimming undergarment for adults and teens.

Applying Structured Teaching Principles to Toilet Training

How to create structure and routine to toilet training for a child with Autism – an article by Susan Boswell and Debbie Gray through the University of North Carolina TEACCH® Autism Program.

Angelman Family Contributions: Diapers

We like Abena Abri Flex diapers. They are available for purchase on Amazon. They are pretty thick and absorbent and are very easy to put on and take off.

Myriah, angel Madeline, age 12, Mutation

Use a diaper doubler inside their diaper at night to help with leakage.

AS Family Member

Tranquility and Tena diapers for children and adults 100 lbs. and more. Many add extra pads.

AS Family Member

Home Delivery Incontinent Supplies (HDIS) is excellent to use (1-800-269-4663) HDIS.com

Pam and Warren, wpnew@windstream.net, angels Jonathan and Andrea, ages 40 and 35, UBE3A Mutation

DISCIPLINE

Angelman Family Contributions: Discipline

My 2 year old Brantley deletion positive knows what the word NO means. I tell him if he's pulling my hair or such "NO" and he shakes his head NO and laughs. I'm pretty sure these kids know more than we think!

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

We use positive reinforcement to promote appropriate behaviors. Punishment techniques have never worked with our son.

AS Family Member

Keep in mind that much of the "negative behaviors" are the result of anxiety, frustration, lack of sleep, illness, etc. Certainly, our angels suffer "fight or flight" at times and this can be traumatic for all. The harder, but more effective strategy, is to whisper reassurance, let them know that everything will soon be OK and that you are there. Patience is the key- even in public when things might become embarrassing. (Most people will be sympathetic.) Remember... "This too will pass." It will take a while- and longer than you want for your child to calm down. He/she might lash out at loved ones with slapping or hitting. Do not take it personally! It is often beyond their control. As much as we try, we cannot imagine what it is like to have Angelman syndrome. NOT resorting to physical discipline or screaming will, in the long run, be more effective and will help you as a parent feel better about

yourself. As Dr. Phil says, "Children need a safe place to land" and you can create that by using comfort rather than anger.

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del+ Class 1

DRINKING: STRAWS

Angelman Family Contributions: Drinking/Straws

Practice drinking from a straw using box drinks with a straw that the liquid can be gently squeezed up the straw working on them trying to suck the liquid up the straw themselves. Once they can drink from a straw practice using oral muscles by drinking thicken liquids from a straw (you can make your own smoothies using applesauce with juice, or yogurt with milk, or pudding with milk).

Debbie Cahill, Occupational Therapist, Rady Children's Hospital, San Diego, CA

My 2 year old Brantley (deletion +) loves The First Years Mickey mouse sippy cups with handles and straws. They're the only cups I can get him to use with straws built into them. He also doesn't ever chew through those straws. The handles makes it easier to hold on to the cup.

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

For angels who have extreme difficulty in swallowing, I think using a soft sprayer is better than using a straw. Not all angels can independently drink from straws. And if we use a water bottle with a straw, and push often the volume of the water is not at the right amount for them to swallow with ease. Their gulp often brings air into their stomach, making them bloat. A small sprayer on the other hand is very easy to handle.

AS Family Member

DROOLING

Most children with Angelman syndrome drool to varying degrees. Parents of older children feel the problem improves as their child gets older, but it does continue occasionally. Some children have had surgery to help lessen drooling. However, several parents have reported that their child's dentist does not recommend surgery since the saliva may actually help prevent cavities as it helps to cleanse the teeth. Scopoderm patches (used to prevent motion sickness) can decrease saliva. Parents report sending wet wipes or baby wash cloths to school or adult programs to help with dryness and germ protection.

Perhaps, a long term option/solution for improving a drooling problem is *occupational therapy*. Since the problem may be rooted in swallowing difficulties and positioning of the tongue, an occupational therapist could offer tips for you and your child's teacher. Request an occupational therapy consultation at your child's IEP meeting.

Angelman Family Contribution: Drooling

We discovered that a large pack of bar cloths (like dish rags from Sam's Club) are great for chewing on, absorbing drool, and then tossing in the washing machine for a fresh start! They get ragged and need to be replaced, but cheap and easy to find! These help calm an angel and are great for "down time". This has made our lives so much more pleasant!!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

E

EDUCATION/SCHOOL

See I- IEP

Back to School Template

Created by an AS parent, download and personalize this template to give to new teachers, therapist, babysitters, etc. to introduce your individual with AS and Angelman syndrome.

Go to: <https://www.angelman.org/resources/back-to-school-template/>

Working with a Child who has Angelman syndrome / The Role of an Early Childhood Special Education Teacher in Kindergarten

Katherine M. Dobbs 4-2017

St. Cloud State University, jkirdobbs2@gmail.com

https://repository.stcloudstate.edu/cgi/viewcontent.cgi?referer=https://www.google.com/&httpsredir=1&article=1019&context=cfs_etds

Individuals with Disabilities Act (IDEA)

Special Education Guide – Profiles of Disabilities

Under the Individuals with Disabilities Education Act (IDEA), there are 13 categories under which a student is eligible to receive the protections and services promised by this law. This site defines each disability as specified by IDEA and explains it in plain English. You can also find common traits and educational challenges associated with each disability, and tips for parents and teachers.

Learning/Assessment Materials

SuperDuper Publications

SuperDuper Publications offers fun learning materials for kids with special needs.

Employability/Life Skills Assessment

Criterion-referenced checklist to assess student's level of performance in the twenty-four critical employability skill areas identified by Ohio's Employability Skills Project.

Literacy and Communication

Dynamic Learning Maps

Learning maps allow students to demonstrate their knowledge, even when they take alternate pathways to achieve that knowledge. These alternate pathways give students more opportunities to show that they can learn challenging content. The DLM® project is guided by the core belief that all students should have access to challenging grade-level content. The new DLM Alternate Assessment System will let students with significant cognitive disabilities show what they know in ways that traditional multiple-choice tests cannot.

Augmentative/Alternative Communication Intervention

Find tips, products and other resources for communication development for children and adults with special needs.

UNC School of Medicine Center for Literacy and Disability Studies

Provides information and resources that promote literacy and communication for individuals of all ages with disabilities. See the plan for Predictable Chart Writing.

SERI

A collection of internet accessible information resources of interest to those involved in the fields related to Special Education.

Understanding Spec Ed FAQ's

Collection of FAQ's about various aspects of Special Education.

Institute of Education Sciences – Find what works

Summarize and compare the evidence of the effectiveness of interventions that address your school or district's needs.

Educational Webinars

<https://www.angelman.org/resources-education/educational-webinars/>

Learn about relevant topics like research updates, clinical development, tips for everyday living, managing symptoms, and many more.

Angelman Family Contributions: Education

Decide what is important to your angel and advocate for that.

AS Family Member

Be realistic about what you expect from your school system. You could spend your entire time fighting for things that your school is not even capable of. This is a marathon... not a race. Conserve your energy!

AS Family Member

Our child has difficulty staying focused and engaged with groups for very long in school. We have found that movement helps him with sensory processing and allows him to function easier. So while sitting during circle time at school, a weighted blanket helps to provide extra input and deep touch pressure, therefore, decreasing his movement needs. We have also heard of people using fidget toys and "move and sit discs". Movement breaks can be important as well.

Desiree, desireemartika@yahoo.com, angel Titus, age 3, Del+

EEG

See S- Seizures



Normal EEG



Angelman syndrome EEG showing typical posterior notched delta activity

<https://www.angelman.org/resources-education/seizure-resources/>

Angelman Family Contributions: EEG

Extended EEG's are miserable for angels. Unless it is truly necessary, we do NOT subject our angel to these anymore.

AS Family Member

ENTERTAINMENT

Angelman Family Contributions: Entertainment

Try something new for entertainment every week, from going to an amusement park, the movies, a kids themed restaurant, a family gathering, the pool, horseback riding, riding a bike/trike, going to a wedding, dancing, playing a board game, walking, cruising the neighborhood, going on vacation, etc, etc. There's a million ways to entertain our Angels, especially during the holidays. Try a new thing every week by creating an event on your calendar or schedule and follow through. Each new thing they try is an opportunity to open doors for independence and for learning. And don't just try it once, try a few times to see if they truly like it or not before moving on to the next new thing. Also, before trying out something new, explain what the surprise for the week is by showing them a video or pictures and talking about the different things they'll see where you will be going. We started this a few years ago and my daughter loves it!

Maria, tinyange15@yahoo.com, angel Vivienne, age 12, UBE3A Mutation

Justin loves to go horseback riding! It started out as therapy when he was 3 but now at 22 he still loves it!! As soon as he gets home from his day program on Tuesdays and Thursdays I have his helmet in my hands and he goes right to my car! This is his totally fulfilling entertainment /fun for him! He smiles the entire time while on his horse Iggy!! I say we are gonna ride Iggy and he lights up with that smile!

Barbara, bbarclay1@verizon, angel Justin, age 22

Our son loves musical performances, the circus, and going to the movies.

AS Family Member

Musicals and any video or image that has dancing seems to be very interesting to my Angel. Especially if the characters are real people and not cartoons.

Tatiana, angel Alina, age 4, UBE3A Mutation

I will never forget the first time I took my daughter to a movie on Thanksgiving Day to see *Frozen*.

Extended family had come to visit, and I didn't want my girl left out, though I wondered if she would make it through the movie. She loved it! After this, we had so much fun as a family planning for another movie outing because this made us all feel so good that EVERYONE could do this. We discovered soon that she liked watching movies at home, too, and so because of her limited eyesight we mounted our TV above a sturdy piece of furniture, so that she can stand and watch her favorite movies and shows. I think this makes our girl feel validated and understood, but we could have so easily second-guessed our decision to take her to the movies, and we would have missed out on this thing she loves so much.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

EPILEPSY AND ANGELMAN SYNDROME

See S-Seizures

Epilepsy Foundation website:

<https://www.epilepsy.com/learn/epilepsy-due-specific-causes/angelman-syndrome>

EXERCISE

Angelman Family Contributions: Exercise

Our angel is food addicted and exercise is very important! It helps him sleep better too!

AS Family Member

Justin loves to go horseback riding! It started out as therapy when he was 3 but now at 22 he still loves it!! As soon as he gets home from his day program on Tuesdays and Thursdays I have his helmet in my hands and he goes right to my car! This is his totally fulfilling entertainment /fun for him! He smiles the entire time while on his horse Iggy!! I say we are gonna ride Iggy and he lights up with that smile!

Barbara, bbarclay1@verizon, angel Justin, age 22

EYE ISSUES

Hypopigmentation, Strabismus and Ocular Albinism

Information from the 2009 document written by Charles A. Williams, M.D; Sarika U. Peters, Ph.D.; and Sarika U. Peters, Ph.D.

[http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20as%202009%203-19-10.pdf)

When AS is caused by the large deletion, skin and eye hypopigmentation usually result. This occurs because there is a pigment gene (the P gene, also termed OCA2), located close to the AS gene, that is also missing [75]. This pigment gene produces a protein that is believed to be crucial in melanin synthesis. Melanin is the main pigment molecule in our skin. In some children with AS, this hypopigmentation can be so severe that a form of albinism is suspected [76]. When AS is caused by the other genetic mechanisms, this gene is not missing and thus normal skin and eye pigmentation is seen. Children with AS who have hypopigmentation are sun sensitive, so use of a protective sun screen is important. Not all AS children with deletions of the P gene are obviously hypopigmented, but may only have relatively lighter skin color than either parent.

Surveys of individuals with AS demonstrate an increased incidence of strabismus. This problem appears to be more common in children with hypopigmentation (as above), since pigment in the retina is crucial

to normal development of the optic nerve pathways. Management of strabismus in AS is similar to that in other children: evaluation by an ophthalmologist, correction of any visual deficit, and where appropriate, patching and surgical adjustment of the extraocular muscles. The hypermotoric activities of some AS children will make wearing of patches or glasses difficult but many are able to accomplish this.

Angelman Family Contributions: Eye Issues

Although there is no evidence linking the development of pterygiums on the corneas of the eyes and Angelman syndrome, I have a strong suspicion that some angels could be more susceptible. My concern is with the affected OCA2 gene in some AS individuals with large deletions. At a very early age, our daughter would go to great lengths to flip around in her stroller and avoid the sun and I believe she had extreme sun sensitivity. Our deletion positive, class 1 daughter with Angelman syndrome developed very aggressive pterygiums in her early 20's. Pterygiums are membranes or "blisters" that grow on the cornea of the eye. People living in sunny, dry climates are especially susceptible, but people anywhere can develop them. Pterygiums begin growing in the corners of the eye and can spread across if left untreated. Our older daughter first noticed the pterygiums on our angel's eyes (we never did) so you might want to ask your angel's doctor to watch for them. Unfortunately, for our angel, scar tissue formed after surgeries to remove the pterygiums, and this necessitated several corneal transplants to restore her eyesight. Research has led us to believe that she has *Ocular Albinism Type 2*. There have been other cases of this in children with AS.

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del+ Class 1

F

FACEBOOK GROUPS

Facebook ASF: <https://www.facebook.com/AngelmanSyndromeFoundation>

To search for Angelman syndrome related Facebook groups:

https://www.facebook.com/search/groups/?q=Angelman%20Syndrome&epa=SEARCH_BOX

Here are links to some groups: Keep in mind that the ASF can only endorse those created and supervised by the ASF. As with any Facebook group, use caution!

- [Angelman Syndrome Foundation](#)
- [\(Angelman syndrome ONLY\) Vagus Nerve Stimulation Therapy](#)
- [ABA for Angelman syndrome](#)
- [Angelman syndrome Families of California](#)
- [Getting In-Sync with Angelman syndrome](#)
- [2018 Angelman syndrome Supporting Greatness](#)
- [2020 Michigan Angelman syndrome Walk](#)
- [A Good Life for those with Angelman syndrome](#)
- [Angelman Connections](#)
- [Angelman directory](#)
- [Angelman Research & Trials](#)
- [Angelman syndrome](#)
- [Angelman syndrome](#)
- [Angelman syndrome & Alternative Health Approach](#)

- [Angelman syndrome Alberta-Saskatchewan Families](#)
- [Angelman syndrome AS Family Members](#)
- [Angelman syndrome Association of WA](#)
- [Angelman syndrome Families - BC](#)
- [Angelman syndrome Families - CANADA](#)
- [Angelman syndrome Families in Indiana](#)
- [Angelman syndrome Families of Arkansas](#)
- [Angelman syndrome Families of Utah](#)
- [Angelman syndrome Find a buddy](#)
- [Angelman syndrome For Parents, Educators, Therapists And Love Ones](#)
- [Angelman syndrome Foundation Walk Portland/Gresham, Oregon](#)
- [Angelman syndrome Imprint Centre Defect \(ICD\) Parent Chat](#)
- [Angelman syndrome India](#)
- [Angelman syndrome Ireland](#)
- [Angelman syndrome Manitoba](#)
- [Angelman syndrome -Nebraska](#)
- [Angelman syndrome NZ](#)
- [Angelman syndrome Seizure Information](#)
- [Angelman syndrome Siblings](#)
- [Angelman syndrome Support](#)
- [Angelman syndrome Support Network of Arkansas](#)
- [Angelman syndrome Tube Feeding Group](#)
- [Angelman syndrome Walk Washington, DC](#)
- [Angelman syndrome, Arizona](#)
- [Angelman syndrome mosaic](#)
- [AngelmanUK Communication & Literacy.](#)
- [AngelmanUK Support & Chat](#)
- [ASF Walk San Diego](#)
- [ASF's Run For A Cure](#)
- [Canadian Angelman syndrome Society Group](#)
- [Chasing Angels - Behaviors in Angelman syndrome](#)
- [Clinically Diagnosed Angelman syndrome Families](#)
- [Cure for Zac - Angelman syndrome](#)
- [Florida Angelman syndrome Parents](#)
- [Foundation for Angelman syndrome Therapeutics United Kingdom](#)
- [Gene Therapy for Angelman syndrome](#)
- [GI Issues in Angelman syndrome](#)
- [Homeschooled Learners with Angelman syndrome](#)
- [International Angelman Day](#)
- [Latter-day Saint Angelman syndrome Families](#)
- [Loghan's Journey with Angelman syndrome](#)
- [Long Island "Angels"](#)
- [Low Glycemic Index Treatment in Angelman syndrome](#)
- [Minnesota Angelman syndrome Parents/Families](#)
- [Minocycline for Angelman syndrome: Results & Progress](#)
- [Pacific Northwest Angelman Syndrome Foundation](#)

- [PODD & Angelman syndrome](#)
- [PODD & Angelman syndrome \(for professionals\)](#)
- [Sharing Causes of Mortality in Angelman syndrome](#)
- [South African Angelman syndrome Group](#)
- [Support For Male Teens, Pre-puberty and Up \(Angelman syndrome\)](#)
- [Testing for Angelman syndrome](#)
- [The Angelman syndrome Group](#)
- [The Loving Angels Support Group](#)
- [UK Angelman syndrome Mums](#)
- [VA & NC Angels \(Angelman syndrome\)](#)
- [VUMC Angelman syndrome](#)
- [Wichita Angelman syndrome Volunteers](#)
- [Wichita Angelman syndrome Walk](#)
- [WINGS for Angelman syndrome](#)

Angelman Family Contributions: Facebook Groups

Be careful!!!! Realize that everything you say in a private group is NEVER truly private.

AS Family Member

They are a great source of support and can ALSO be a source of depression when you read all kinds of negative things in FB groups.

AS Family Member

FACTS ABOUT ANGELMAN SYNDROME

A MUST READ!

Use the following link to read the 2009 document written by Charles A. Williams, M.D; Sarika U. Peters, Ph.D.

[http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20as%202009%203-19-10.pdf)

FAMILY-RELATED TOPICS

See P- Parents Health

See M- Marriage and Relationships

See S- Siblings

Angelman Family Contributions: Family

Family members will have A LOT of opinions. Trust your gut and ALWAYS do what is best for your angel and your immediate family.

AS Family Member

Remember that moms and dads deal with AS differently!

AS Family Member

FEMALE ISSUES

Angelman Family Contributions: Female Issues

Not all angel girls need to get on birth control immediately. Wait a few cycles and see what's best for her. It's not about you!

Norma, angel Katarina, age 20, Del +

When our daughter was older, we consulted with a gynecologist about birth control methods. As they say, unless you are with your child 24/7, there is always a risk- as awful as that seems. The method we chose was a low dose birth control pill. Under the directions of the doctor, we always skip the last "placebo" week and begin a new pack of pills. Consequently, our daughter does not bleed and this has improved her life tremendously. This wonderful doctor even came to the hospital when our daughter was having eye surgery under general anesthesia and performed a pelvic exam. An added benefit of birth control pills is reducing her risk of ovarian cancer.

AS Family Member

FINANCIAL ASSISTANCE

ASF Family Fund

Families apply for funds to assist in gaining access to resources that are needed to improve the quality of life for an individual with Angelman syndrome.

Joyful Journey Mom

Ultimate list of grants and resources for families with special needs

Go Fund Me

Offers the ability to raise funds. Create a campaign, share it with friends and family and collect donations.

Help Me Fly Inc.

Help Me Fly Inc. was started by the parent of an AS individual. The organization helps families gain access to adaptive equipment that they normally could not have access to without some type of outside help.

Social Security Benefits for Children with Disabilities (2014)

Information for the parents, caregivers or representatives of children younger than age 18 who have disabilities that might make them eligible for Supplemental Security Income (SSI) payments. It is also for adults who became disabled in childhood and who might be entitled to Social Security Disability Insurance (SSDI) benefits. Find information about how to apply for benefits, employment programs for young people with disabilities, Medicaid, Medicare and children's health insurance programs.

Jill Fox Memorial Fund, Inc.

Maryland Residents

The Jill Fox Memorial Fund provides grants to individuals with unmet medical and healthcare needs not paid for by medical insurance, government agencies, health associations, community organizations, public or personal resources.

The Enterline Foundation

Established to provide financial resources to improve the lives of individuals with special needs – who

are intellectually disabled and developmentally disabled, The Enterline Foundation is committed to delivering resources to help this under-served and often overlooked part of our community. The Enterline Foundation provides financial support to organizations that in turn provide direct services to adults and children with intellectual and developmental disabilities.

Urban League of Rochester Developmental Disabilities Division

New York residents

The Family Reimbursement Program provides financial assistance to families caring for individuals with developmental disabilities. Funds assist in preventing out-of-home placements and enhance the quality of family life.

The Chatham County Department of Social Services

Chatham County, North Carolina residents

Mission: The Chatham County Department of Social Services will protect vulnerable children and adults; strengthen and preserve families; provide access to health care and better nutrition; and promote economic stability while encouraging personal responsibility.

Adult Services: Provides an array of services and financial assistance so that the aging and developmentally disabled adults can either remain in their homes as long as possible or can reside safely in an appropriate facility.

The Agency for Persons with Disabilities (APD)

Florida (Dade and Monroe counties) residents

The Agency for Persons with Disabilities (APD), in partnership with a Miami dentist opened the Bay Harbor Complete Dentistry, which provides children and adults with developmental disabilities in Dade and Monroe counties who are unable to find dental care. APD eligible individuals, including those on the waiting list for waiver services, may receive cleanings, screenings, x-rays, and other specialized services at reduced cost or with financial assistance. If you live in Dade or Monroe Counties, contact the APD Area 11 office in Miami at 305-349-1478 to make an appointment.

Easter Seals

East Georgia residents

The Champions for Children Program is designed to primarily assist children and families who do not meet the eligibility requirements for the TEFRA/Katie Beckett Medicaid program. The program will provide direct financial assistance and support services for Georgia's medically fragile and special needs children and their families. An Easter Seals Champions for Children Coordinator will work directly with families to determine eligibility, identify service needs as described by the family and connect them with resources and services. In addition, coordinators will also promote the program and raise awareness of other resources and services in the community. Families of medically fragile and special needs children across Georgia are able to apply for and receive direct financial assistance and support services from the state-wide nonprofit collaborative to help cover the costs of caring for their children at home.

Friends of Disabled Adults & Children

Stone Mountain, Georgia residents

Non-profit organization providing free wheelchairs and home healthcare equipment for the disabled: Other services include ramp building, vehicle lift installation and more.

Alabama Family Trust

Birmingham, AL residents

Many Americans depend upon governmental entitlements such as Medicaid and SSI to provide for their financial needs. Many of these individuals have mental or physical disabilities that substantially limit their major life activities. However, the financial resources provided by Medicaid and SSI are often insufficient to cover the day-to-day living expenses of those that depend upon these programs. This is particularly true of those with disabilities, who must also face the costs of increased medical bills and the need for special medical equipment and supplies. The Alabama Family Trust (AFT) provides trust services for persons with such disabilities. The organization was created by the State of Alabama Legislature in 1994 to encourage, enhance, and foster the provision of medical, social, or other supplemental services for persons with a mental or physical impairment (See Code of Alabama 1975 Chap. 38-9B). The AFT is a 501 (c)(3) general non-profit organization established for the purpose of administering special needs trusts for the disabled.

FINANCIAL FEDERAL RESOURCES

Medicaid Waivers by State

Find information and help for Medicaid waivers for every state.

Social Security – SSI for Children

Information for the parents, caregivers or representatives of children younger than age 18 who have disabilities that might make them eligible for Supplemental Security Income (SSI) payments. It is also for adults who became disabled in childhood and who might be entitled to Social Security Disability Insurance (SSDI) benefits. Find information about how to apply for benefits, employment programs for young people with disabilities, Medicaid, Medicare and children's health insurance programs.

Disability Legal Help

As a social security disability law firm, we have been helping people successfully process disability claims for over 30 years. Our lead disability attorney Fred J. Fleming's knowledge, experience and understanding of the claims process has resulted in a very high success rate. Statistically, your chances of a successful result are much higher with representation.

National Guardianship Association Affiliates by State

Find information about guardianship laws by state. Each state has different laws and procedures for becoming a guardian, government financial benefits, future planning and more.

FINANCIAL PLANNING

<https://www.angelman.org/resources-education/resources/financial-planning/>

CA Families – IRS Medicaid Waiver Information

Please check with your tax advisor to see if you qualify for a tax credit or refund per the following IRS notice. <https://www.irs.gov/pub/irs-drop/n-14-07.pdf>

Protected Tomorrows, Inc.

Protected Tomorrows, Inc., enhances the lives of people with developmental disabilities through comprehensive life planning. Its nationwide network of Advocates creates Future Care Plans™, which

include the educational, residential and financial needs of the individuals. Protected Tomorrows brings peace of mind as it continuously finds progressive solutions to family concerns.

Merrill Lynch

Special Needs Financial Services at Merrill Lynch provides tools and services for clients with disabilities and their families. Our dedicated network of trained financial professionals can help you address the lifetime financial needs of a disabled child or loved one (see “Great Challenges, Greater Achievements”). We also provide communications accessibility tools for clients with hearing or visual impairment as well as medical bill management and claims-filing services for individuals with long-term health-care needs.

MassMutual SpecialCare (previously MetLife MetDESK)

Planning for the future of your child or other dependent with special needs can be a complicated process. It’s important, however, to plan for their care and continued quality of life when you are no longer around. Trained professionals can help guide you through the process and help you achieve the peace of mind that comes when you know you have planned appropriately.

National Guardianship Association Affiliates by State

Find information about guardianship laws by state. Each state has different laws and procedures for becoming a guardian, government financial benefits, future planning and more.

Angelman Family Contributions: Financial Planning

It is never too early to begin financial planning for your child’s future.

AS Family Member

Don’t expect our government to provide everything your angel will ever need. Try to be financially wise so you always have a little extra to provide things that government assistance won’t.

AS Family Member

G

GASTROINTESTINAL ISSUES AND ORAL MOTOR BEHAVIORS

See A- Acid Reflux

See C- Constipation

See G- GERD

See N- Nausea and Vomiting

http://www.angelman.org/wp-content/uploads/2015/11/facts_about_as_2009_3-19-10.pdf

Feeding problems are frequent but not generally severe and usually manifest early as difficulty in sucking or swallowing [67--69]. Tongue movements may be uncoordinated with thrusting and generalized oral-motor incoordination. There may be trouble initiating sucking and sustaining breast feeding, and bottle feeding may prove easier. Frequent spitting up may be interpreted as formula intolerance or gastroesophageal reflux. The feeding difficulties often first present to the physician as a problem of poor weight gain or as a “failure to thrive” concern. Infrequently, severe gastroesophageal reflux may require surgery.

Angelman children are notorious for putting everything in their mouths. In early infancy, hand sucking

(and sometimes foot sucking) is frequent. Later, most exploratory play is by oral manipulation and chewing. The tongue appears to be of normal shape and size, but in 30-50%, persistent tongue protrusion is a distinctive feature. Some have constant protrusion and drooling while others have protrusion that is noticeable only during laughter. Some infants with protrusion eventually have no noticeable problem during later childhood (some seem to improve after oral-motor therapy). For the usual AS child with protruding tongue behavior, the problem remains throughout childhood and can persist into adulthood. Drooling is frequently a persistent problem, often requiring bibs. Use of temporary medications such as scopolamine to dry secretions usually does not provide an adequate long term effect. Surgical procedures to ameliorate drooling are possible [70] but apparently rarely used in AS.

NORD <https://rarediseases.org/rare-diseases/angelman-syndrome/>

Feeding difficulties may be treated by modified breast feeding methods and by means such as special nipples to assist infants with a poor ability to suck. Gastroesophageal reflux may be treated by upright positioning and drugs that aid the movement of food through the digestive system (motility drugs). Surgical tightening of the valve that connects the esophagus to the stomach (esophageal sphincter) may be required in some cases. Laxatives may be used to treat constipation.

GENETIC COUNSELING

https://www.peds.ufl.edu/divisions/genetics/programs/angelman_syndrome/genetic_counseling.htm

Do not use the following aspects in lieu of consulting with a genetic counselor. These are aspects to consider in understanding AS genetic risk:

1. Common chromosome deletion:

More than 98% of the chromosome deletion instances occur by a spontaneous event and thus they are not inherited; the recurrence risk is <<1% for these families. However, 1-2% of deletions occur because of an inherited abnormality in the maternal chromosome 15, such as a balanced chromosome translocation. Another very small group (e.g., only a few cases reported in the literature), can have AS due to a very small, maternally inherited chromosome deletion that involves a small area around and including the UBE3A gene. For these cases, the maternal recurrence risk is increased depending on the type of abnormality present. Chromosome study of the mother, including FISH, helps rule out inherited chromosome 15 abnormalities.

2. Paternal uniparental disomy (UPD):

More than 99% of patUPD cases occur as an apparent spontaneous, non-inherited, event. If an individual has AS due to patUPD and has a normal karyotype, a chromosomal analysis of the mother should nevertheless be offered in order to exclude the rare possibility that a Robertsonian translocation or marker chromosome was a predisposing factor (e.g., via generation of maternal gamete that was nullisomic for chromosome 15, with subsequent post-zygotic "correction" to paternal disomy).

3. Imprinting Center (IC) Defect:

There are two types of IC defects: deletions and non-deletions. Non-deletion events do not appear to be inherited and have a <1% recurrence risk. Most deletions are not inherited but a significant proportion of them are (i.e., maternally inherited), and these confer a 50% risk for recurrence.

4. UBE3A mutations:

UBE3A mutation can either occur spontaneously (e.g., not inherited and with no increased recurrence risk) or be maternally inherited and have a 50% risk of recurrence (see below for imprinting inheritance).

5. Individuals with no known mechanism (all 4 above mechanisms have been eliminated):

For parents of AS individuals who have apparent normal genetic tests (no evidence for deletion, imprinting defect, UPD or UBE3A mutation), and thus their children are only clinically diagnosed, it is not known what the recurrence risk is. An increased risk seems likely but probably does not exceed 10%.

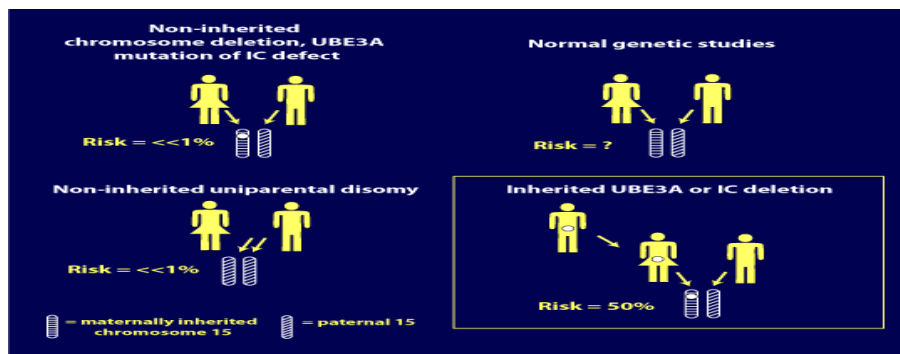
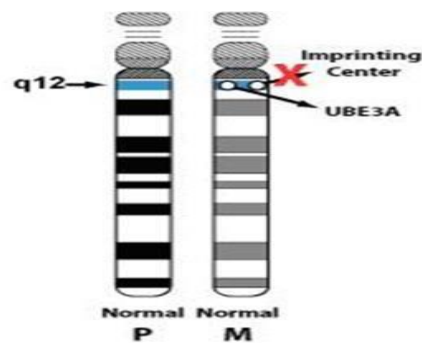
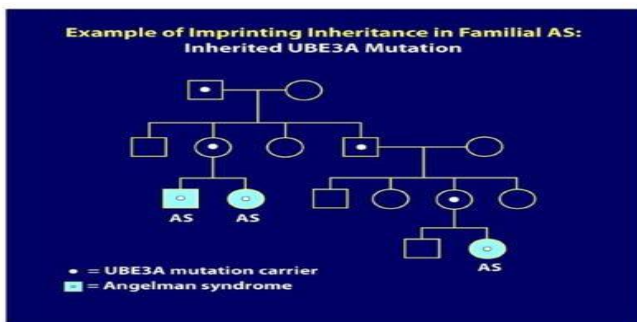
6. Germ cell mosaicism:

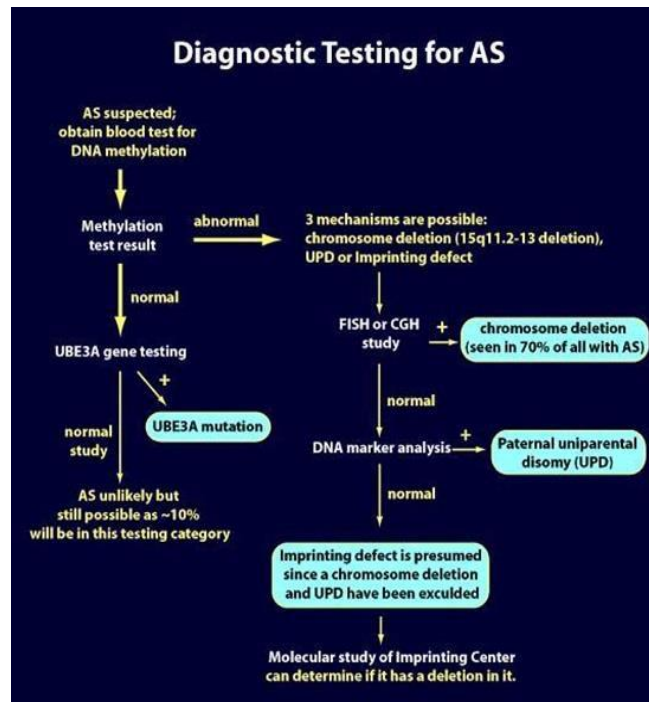
This term refers to a phenomenon in which a genetic defect is present in the cells of the gonad (ovary in the mother's case) but not in other cells of the body. This occurrence can lead to errors in risk assessment because a genetic test, for example on a mother's blood cells, will be normal when in fact a genetic defect is present in the germline cells of her ovary. Fortunately, germ cell mosaicism occurs very infrequently. Nevertheless, it has been observed in AS caused by the mechanisms of large chromosome deletion, Imprinting Center deletion and UBE3A mutation.

7. Imprinting inheritance:

UBE3A mutations and Imprinting Center deletions can exhibit imprinting inheritance wherein a carrier father can pass on the genetic defect to his children without it causing any problems, but whenever a female passes this same genetic defect on to her children, regardless of the sex of her child, that child will have AS. The pedigree diagram below illustrates imprinting inheritance. Here, AS has only occurred after a carrier mother passed on the gene defect (for example as in the two siblings with AS pictured on the left lower part of the pedigree). In addition, a distant cousin in this family also has AS due to the imprinting inheritance. In the diagram, individuals with the light blue circles or squares have AS but everyone else in the family is clinically normal. The white dots represent asymptomatic, normal carriers of the AS mutation. When an AS genetic mechanism is determined to be inherited, genetic testing of family members can usually identify carriers of the gene defect. As you might imagine, professional genetic counseling is advised in these situations.

GENETICS 101





The following is Dr. Charles Williams' presentation at the 2014 CASS conference: [Infancy to Adulthood: Understanding Angelman syndrome After Five Decades](#)

Angelman syndrome (or AS) is caused by the lack of function of one specific gene, called UBE3A. To understand this, we first need to understand how chromosomes and genes work together to allow our brains to function.

A typical person has 46 chromosomes inside every cell in their body. We inherit 23 chromosomes from one parent and 23 from the other. Chromosomes are like the instruction manuals containing all the detailed information our cells need in order for our brains and bodies to grow and function. Chromosomes are numbered like volumes in a set of encyclopedias. We inherit one copy of each chromosome from each parent, giving us two copies of each chromosome. We all have two copies of the chromosomes numbered from 1 to 22, plus a sex chromosome inherited from each parent. Our genes are like the individual topics in the set of encyclopedias. Different genes are located in each chromosome the way that information might be organized in an encyclopedia. Scientists only understand a fraction of how our genes help cause good health or disease and exactly how each gene functions.

It is likely that we all have a number of genetic errors in our chromosomes. This is one reason why it is good that we have two of each chromosome to draw from: if one chromosome has an error, our cells can easily get the information they need from the second chromosome. Nature has provided us with a great back-up system to ensure our cells always have the instructions they need for our brains and bodies to function even when there are inevitable errors in our chromosomes.

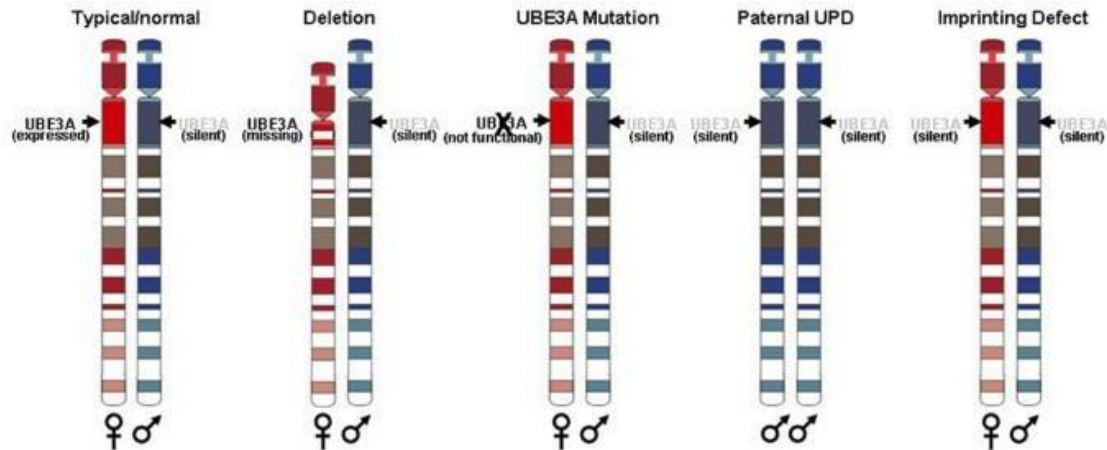
The important chromosome for Angelman syndrome is number 15. This chromosome has a region that is "imprinted." Imprinting means that some genes on the chromosome are "turned on" or "turned off" depending on which parent contributed the chromosome. Going back to the set of encyclopedias,

imagine that we have two versions of the 15th volume, but a chapter in the volume inherited from the father has been washed out and the “pages” are now blank. Similarly, a different chapter in the volume from the mother is washed out and the pages appear blank. Imprinting occurs at the time of conception as part of the normal development of the fetus and each of us have regions on some of our chromosomes that are imprinted, meaning that only one parent’s genetic information is available to our cells as instructions on how to grow and develop. In imprinted regions of our chromosomes, only one parent’s information is accessible to cells in the brain and body, so there is no back-up system if there is an error in the remaining chromosome.

There are many mechanisms that cause what is known as Angelman syndrome. The most common is a “deletion.” If the 15th chromosome is like a volume in an encyclopedia, with a special chapter that is only in the volume inherited from the mother, then imagine that chapter has been torn out. This is a bit similar to how most people with Angelman syndrome have a deletion in their chromosome 15. There is a missing chapter in their volume so the brain is missing some of the instructions it needs to grow and develop. The father’s chapter is present but was “washed out” due to the natural process of imprinting so the pages appear blank and the cells in the brain can’t access the information. Other individuals with Angelman syndrome have a mutation in the UBE3A gene. This is similar to having a misspelling in the important chapter that is only present on the chromosome inherited from the mother. This misspelling is so severe that it makes much of the chapter illegible to cells in the brain. Again, the end result is that the brain lacks important information to learn and develop. Some people with Angelman have uniparental disomy or UPD. In these cases, the individual inherited two copies of this “volume 15” from the father and no copy from the mother. Again, the information that is only in the volume inherited from the mother is missing and both copies from the father are blank or “washed out”.

A small number of individuals with Angelman have an imprinting centre (IC) defect on the maternal chromosome. There is a region on the 15th chromosome that helps the chromosome decide whether the chapters of information are accessible or “washed out”. When there is a defect in the maternal IC, the chapters that are normally accessible become inaccessible. Even though the “volume 15” is complete and normal, the cells in the brain can’t access the information that they need.

Lastly, a few individuals have all or most of the symptoms of Angelman syndrome, but every test of their genes turns out normal. In many of these cases, it is likely that we just don’t yet have the technology and expertise to understand what is wrong, but additional research is likely to reveal new causes of AS, which will help these families understand what is causing Angelman syndrome in their loved one(s). Scientists are still trying to understand what exactly the UBE3A gene does. All we know for sure is that UBE3A is vital to how the brain develops and controls speech, movement and learning. When the UBE3A gene is blocked from functioning normally, the individual has Angelman syndrome. UBE3A is a gene that is also being investigated for a role in autism and genetic disorders like Isodicentric 15. Research into future treatments and cures for Angelman relies on finding a way to make the UBE3A present in the paternal “volume” available to the brain. Essentially, if we think of the genetic information on the father’s chromosome as being “washed out” and invisible, scientists are searching for ways to make the data reappear so that the brain can access the information and individuals with AS can develop to their fullest potential.



The illustration of Chromosome 15 highlights in red the section that contains the UBE3A gene and is often deleted from the maternal chromosome in Angelman syndrome.

The illustration on the right shows the different mechanisms that cause Angelman syndrome. The red chromosomes represent the chromosome inherited from the mother while blue represents the father. The first example shows a normal pair of “volume” 15s with the father’s chromosome chapter with UBE3A “washed out” or silent, while the mother’s chapter is active and UBE3A is expressed, making the genetic instructions in that area available to the cells in the brain. The remaining examples illustrate the known mechanisms that cause Angelman syndrome by making the mother’s genetic information inaccessible to the cells in the brain. For example, in deletions, the UBE3A gene is completely missing as that “chapter” has been “torn out”. In UBE3A mutations, the UBE3A gene is “misspelled” which makes this copy of UBE3A non-functional.

Angelman Family Contributions: Genetic Counseling

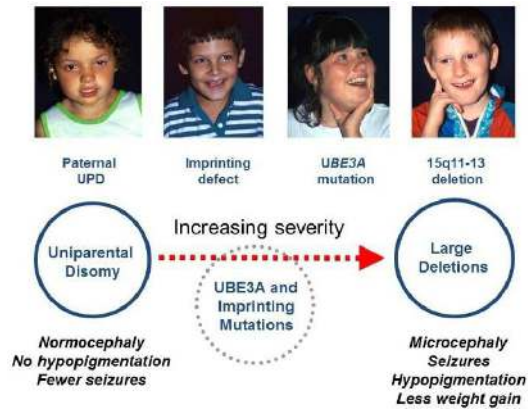
When your child is diagnosed with AS, and if the child has the UBE3A mutation, make sure the mother is tested to see if she is a carrier. (That is if you want to have more children.) That way you will know if there is a chance your next child could have AS.

AS Family Member

GENOTYPE TO PHENOTYPE RELATIONSHIP

Does the type of genetic cause of Angelman syndrome make a difference in development?

(Genotype to Phenotype Relationship) Dr. Charles Williams used this illustration (see next page) in his genetic presentation at the 2014 CASS (Canadian Angelman Syndrome Society) conference.



<https://www.ncbi.nlm.nih.gov/books/NBK1144/>

All genetic mechanisms that give rise to AS lead to a somewhat uniform clinical picture of severe-to-profound intellectual disability, movement disorder, characteristic behaviors, and severe limitations in speech and language. However, some clinical differences correlate with genotype [Fridman et al 2000, Lossie et al 2001, Varela et al 2004, Tan et al 2011, Valente et al 2013]. These correlations are broadly summarized below:

- The 5- to 7-Mb deletion class results in the most severe phenotype with microcephaly, seizures, motor difficulties (e.g., ataxia, muscular hypotonia, feeding difficulties), and language impairment. They also have lower body mass index compared to individuals with UPD or imprinting defects [Tan et al 2011]. There is some suggestion that individuals with larger deletions (e.g., BP1-BP3 [class I; ISCA-37404] break points) may have more language impairment or autistic traits than those with BP2-BP3 (class II; ISCA-37478) break points [Sahoo et al 2006] (see Figure 2).
- Individuals with UPD have better physical growth (e.g., less likelihood of microcephaly), fewer movement abnormalities, less ataxia, and a lower prevalence (but not absence) of seizures than do those with other underlying molecular mechanisms [Lossie et al 2001, Saitoh et al 2005, Valente et al 2013].
- Individuals with IDs or UPD have higher developmental and language ability than those with other underlying molecular mechanisms. Individuals who are mosaic for the nondeletion ID (approximately 20% of the ID group) have the most advanced speech abilities; they may speak up to 50-60 words and use simple sentences.
- Individuals with chromosome deletions encompassing *OCA2* frequently have hypopigmented irides, skin, and hair. *OCA2* encodes a protein important in tyrosine metabolism that is associated with the development of pigment in the skin, hair, and irides (see Oculocutaneous Albinism Type 2). However, other factors in addition to haploinsufficiency of *OCA2* appear to account for the relative hypopigmentation in individuals with AS, as *UBE3A* has now been shown to modulate melanocortin 1 receptor (MC1R) activity in somatic tissues.

GERD

See A- Acid Reflux

What is Gastroesophageal Reflux?

Gastroesophageal refers to the stomach and esophagus. Reflux means to flow back or return.

Gastroesophageal reflux disease, or GERD, is a digestive disorder that affects the lower esophageal

sphincter (LES), the ring of muscle between the esophagus and stomach. Gastroesophageal reflux is the return of the stomach's contents back up into the esophagus.

In normal digestion, the lower esophageal sphincter (LES) opens to allow food to pass into the stomach and closes to prevent food and acidic stomach juices from flowing back into the esophagus. Gastroesophageal reflux occurs when the LES is weak or relaxes inappropriately, allowing the stomach's contents to flow up into the esophagus.

A few examples of potential medications are: Nexium, Protonix, Prilosec, and Prevacid. *** Always get advice from the doctor!

GERM CELL MOSAICISM

This term refers to a phenomenon in which a genetic defect is present in the cells of the gonad (ovary in the mother's case) but not in other cells of the body. This occurrence can lead to errors in risk assessment because a genetic test, for example on a mother's blood cells, will be normal when in fact a genetic defect is present in the germline cells of her ovary. Fortunately, germ cell mosaicism occurs very infrequently. Nevertheless, it has been observed in AS caused by the mechanisms of large chromosome deletion, Imprinting Center deletion and UBE3A mutation.

GIFTS

See T-Toys

Angelman Family Contributions: Gifts

Be generous with care staff. We like to give gifts to let them know they are appreciated. Even a nice handwritten card or handmade gift is wonderful. They are underpaid for the job they do.

AS Family Member

We tend to be very generous with gifts to our son's caregivers. They work hard and are underpaid!!

AS Family Member



Our son always enjoyed soft or sturdy talking toys and stretchy items, bath toys and also blocks that can be put in and out of containers, like a mailbox shape sorter, or simply Legos and large plastic containers!
Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

My tip is to make unwrapping one of the highlights of the moment. My daughter loves slowly pulling apart the paper and finding the gift inside. When everyone else is done with Christmas unwrapping, we all love to watch her take her time with a big smile as she slowly unwraps each gift.

Sarah, bnamommysfun@yahoo.com, angel Lily, age 14, Del+

GOVERNMENT ASSISTANCE

Resources and Services specific to State Waivers, Government Assistance, Insurance and Advocacy.

Contact **Dr. Eric Wright**, ASF Family Resource Team via a form on the ASF website.

<https://www.angelman.org/resources-education/asf-family-resource-team/>

Eric's experiences as an elementary school guidance counselor and teacher at the university level in Education and Human Development make him the ASF's Government/Insurance/Advocacy guru. Along with helping AS families navigate through the red tape of waivers, assistance, and insurance, he also serves as consultant on the Bureau of Maternal Child Health/Pacer Program as well as on the Medicaid Technical Advisory Council and Commonwealth Council on Developmental Disabilities.

Eric and his wife Debbie have three children: Ella, Elsie (Del+), and Ethan. The Wrights love traveling, horseback riding, and swimming together. Together Eric and Debbie continue to seek out resources to best serve other AS families.

GUARDIANSHIP

<https://www.guardianship.org/advocacy/guardianship-in-the-states/>

The map provides information about state guardianship associations across the country. Click on individual states to learn more.

Adult Guardianship:

Watch the ASF Educational Webinar on *Guardianship*, presented by Dr. Eric Wright.

<https://www.youtube.com/watch?v=qhdj-2jRaSc&feature=youtu.be>

H

HAIR CUTTING

Angelman Family Contributions: Hair Cutting

I stand my son in front of the bathroom sink – filled with water, of course – and he becomes so distracted splashing and playing that if I work fast, he barely notices that I am trimming his hair.

AS Family Member

I have a “three step hair cutting plan”: 1) Put the “torture” chair in front of the TV and play a favorite video; 2) Keep a stock of favorite snacks on hand. Food is the great distracter; and 3) If all else fails, get a sibling to entertain your child! This doesn’t make the process easy... just easier!

AS Family Member

I cut my adult son’s hair and it is fun. My tip to avoid hair getting all over is to put a slick windbreaker jacket on backwards with the snaps or zipper in back. The hair easily falls off. Then I put baby powder on Frank’s skin to help the hair brush off. Also use the baby powder after a walk on the beach for legs, hands, and feet to help the sand fall right off.

Daniele, angel Frank

My tip here may not work for everyone, but I cut my girl’s hair myself. I’m not trained, but I have figured out that when I’m at home and can have someone help distract her, I do better than many who have tried at a salon to cut her hair. The main reason is that I have plenty of time and don’t feel rushed to finish. Get a good set of shears and maybe give it a go.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

HAIR PULLING

Angelman Family Contributions: Hair Pulling

My daughter goes through times of intense hair pulling, most recently leaving a small bald spot on her pretty head. Here’s what we do when it starts and lingers more than a couple days: 1. We take her to a pediatrician to rule out infections, and we have him check her ears, nose, throat, and tummy. 2. We often take her to a dentist to rule out a cavity. 3. If these check out, we try Advil for a possible headache. The last episode was so bad and none of the above helped. So we cut her hair very short (she looks very cute), and we changed shampoo (Aveeno Sensitive Scalp). These two things have helped and for now have curbed her hair pulling. We are guessing her head may have been incredibly itchy. Final note: I always look at the hair-pulling as a means to communicate that she is uncomfortable. I do not treat it as a negative attention-seeking behavior. I think of my other verbal kids and wonder what they might be reacting to if they were pulling their hair and crying.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

Tickle your angel for a quick release! This is a quick and effective technique when your angel is grasping someone’s hair in public!

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del+

HISTORY OF ANGELMAN SYNDROME AND THE ASF

<https://www.angelman.org/about/history/>

1965: Discovery of AS

Dr. Harry Angelman, an English physician at Warrington General Hospital, published a research paper that first described children with characteristics of Angelman syndrome. At that time, he referred to the disorder as “Happy Puppet Syndrome.” The condition was considered to be extremely rare at that time, and many physicians doubted its existence. This discovery, in theory, was the birth of AS.

1982: AS Diagnoses

Dr. Charles Williams and Dr. Jaime Frias of the University Of Florida College Of Medicine saw their first patients whom they believed had “Happy Puppet Syndrome” (now known as Angelman syndrome) based on Dr. Angelman’s past observations. It became clear that the condition was more common than previously thought. Dr. Williams and Dr. Frias proposed to change the name of this disorder to Angelman syndrome, in honor of Dr. Harry Angelman’s initial findings in 1965.

1987: Genetic Marker Identified

Ellen Magenis, physician at the Oregon Health Science Center, discovered a genetic “marker” for AS – a missing genetic code on a tiny portion of chromosome 15.

1989-1998: Volunteers Managed the ASF

1991: First Major Gift:

Former Major League Baseball pitcher, Bryan Harvey, donated \$20,000 to the ASF for AS research and family support.

1992: ASF Incorporated

The ASF was officially incorporated, which was the start of 25 years of commitment to research to discover AS treatments and a cure, and supporting families.

1992: Fresno, CA Mini-Conference

Pioneering California families gather “in the middle” and host a two day mini-conference at a local elementary school. Dr. Hutchinson, a local neurologist speaks along with other physicians.

1993: First ASF Conference in Orlando, Florida

The ASF hosts its first official Family Conference as an incorporated organization, with hundreds of families attending. Dr. Harry and Audrey Angelman attend the conference and are honored to meet “their children”. An ASF Board of Directors was elected.

1994: California families gather at Anaheim Stadium in Anaheim, CA

Bryan and Lisa Harvey hosted the event. The families attended a *California Angels* ballgame and relief pitcher, Bryan Harvey sealed the victory! Before the game, the families were invited into the dugout to meet the players where there were autographs signed and photos taken!

1995: Second ASF Conference in Colorado Springs, Colorado

There was an 80th birthday celebration for Dr. Angelman, but sadly he was unable to attend after being diagnosed with cancer.

1996: Dr. Angelman passes on August 8, 1996. He was born on August 13, 1915.

1996: Facts About AS, Dr. Charles Williams, Dr. Sarika Peters, Dr. Stephen Calculator

Facts About Angelman syndrome was published: the first-ever comprehensive resource for families for everything known about AS at that time in history.

1996: First Clinical Trial

The ASF formed its *Scientific Advisory Committee*, awarding ASF's first-ever research grant to Dr. Joseph E. Wagstaff to investigate disturbed sleep patterns. This was the first clinical treatment trial in AS research, [*using melatonin for sleep disturbances*](#).

1997: Mutations Discovered

Dr. Arthur Beaudet discovered mutations in the UBE3A gene as the cause of AS. This discovery quickly led to the development of animal models and active neuroscience research aimed at discovering how abnormalities of UBE3A cause impairment in neural development.

1997: First Scientific Symposium and Family Conference Seattle, Washington

The ASF holds the first Scientific Symposium in conjunction with the 1997 Family Conference in Seattle, Washington. This two-day symposium is a chance for leading researchers, scientists and doctors to discuss the latest research activities in the world of AS. In 2008, the Scientific Symposium became an annual event.

1998-2003: ASI Association Management Company, ASF

1999: Audrey Angelman passes. February 2, 1936 – August 15, 1999

1999: Inaugural Walk

The ASF hosts its inaugural [ASF Walk](#), where more than 200 people walked five miles in Naperville, IL and raised nearly \$25,000. The Walk now has more than 10,000 annual participants at nearly 50 sites across North America, including Canada and Mexico.

1999: Harry & Audrey Angelman Award

The first Harry and Audrey Angelman Award for Outreach and Education was announced by the ASF, honoring the Angelmans for their immense contributions to our community.

2000: ASF Website Launch

2000: Claudia Benton Research Award

The Dr. Claudia Benton Award for Research was announced by the ASF to honor Claudia Benton, who passed away in 1998. The award recognizes those with a strong commitment to advancing the scientific knowledge and understanding of AS or makes a significant impact on the lives of individuals with AS through research.

2003: Eileen Braun named Executive Director of the ASF

Sheila Wenger assisted Eileen with the ASF Walk, Special Events Coordinating, Member Services, and the ASF Biennial Conferences.

2004-Present: ASF is Self-Managed

2011: Paternal UBE3A Activated

Dr. Ben Philpot proved the paternal copy of UBE3A can be activated, a huge breakthrough in AS research. As discovered by Dr. Art Beaudet in 1996, AS is caused by a deficiency of UBE3A, so Dr. Philpot's finding was a very significant and promising step towards better treatments and a cure.

2007: \$1 Million Milestone

The ASF achieved a major milestone in AS research by hitting \$1 million invested.

2009: \$1 Million in Research

The ASF funded more than \$1 million in research in one year for the first time. *More than \$1 million in grants were awarded to six principle investigators focusing on AS research.*

- Benjamin Philpot, PhD – University of North Carolina Chapel Hill
- John Marshall, PhD – Brown University, Providence, RI
- Eric Klann, PhD – New York University
- Peter Howley, MD – Harvard Medical School
- Yong-Jui Jiang, MD, PhD – Duke University, Durham, NC
- Scott Dindot, PhD – Texas A&M University, College Station, TX

2012: AS Clinics Established

2013: International Angelman Day

International Angelman Day was celebrated for the first time to help raise awareness for AS across the world. That year alone, ASF Facebook posts were shared more than 1,000 times, introducing AS to thousands of people for the very first time.

2013: iPads for AS

The iPads for AS initiative enabled the ASF to award 100 iPads to individuals with AS for communication aids.

2013: \$5 Million Milestone

ASF funded research reached \$5 million invested. In 2013, the ASF committed over \$200,000 to fund Angelman syndrome research putting the cumulative amount invested to over \$5 million!

2014: ASOs Activate Paternal UBE3A

Dr. Art Beaudet builds on Dr. Philpot's paternal UBE3A activation discovery, using antisense oligonucleotides (ASO) to activate paternal UBE3A. This successful research moved us even closer to a therapeutic.

2015: AS Clinic at Mayo Clinic

Mayo Clinic invested in AS research by opening its own *AS Clinic in Rochester, Minn.*, on November 30, 2015, marking the third AS Clinic available to families in the U.S.

2015: Communication Training Series

The *ASF Communication Training Series* started, a program designed for families to help their individual with AS reach his or her greatest communication success. To date, more than 53,110 people have participated in these webinars.

2016: Center for Courageous Kids

The ASF organized and funded its first Center for Courageous Kids (CCK) outing, a fully-inclusive camping experience for 30 AS families. The ASF funded a second outing in April 2017.

2017: Biomarker Identified for Clinical Trials

ASF-funded research, published in the *Journal of Neurodevelopmental Disorders*, identified that delta—a frequency of brain rhythms identifiable by EEG scans—can serve as a reliable biomarker for pre-clinical and clinical trials in Angelman syndrome, one of the first biomarkers to be established in AS research. Biomarkers play a critical role in determining whether a potential therapeutic is effective, and this study gave the AS research community a viable tool to measure success of many trials!

2018: \$10 Million Milestone

ASF funded research reached \$10 Million invested. With so many advances being made in Angelman research by talented, dedicated professionals, the ASF is striving to have \$10 million invested in AS research during the year 2018.

2019: Amanda Moore becomes the first CEO of the Angelman Syndrome Foundation

2019: ASF Family Fund

The ASF Family Fund was created. This allows families to apply for funds to gain access to resources needed to improve the quality of life for an individual with Angelman syndrome. Applications for funding are open twice a year in October and April.

HUMOR

Angelman Family Contributions: Humor

Humor is a huge coping mechanism because if we did not laugh at some of the things our angel does, we would probably cry.

AS Family Member

***Appeared in the 1999 edition, *Angelman syndrome from A to Z*. Artist P.J. has a sibling with Angelman syndrome.



In church several years ago, the family went to the altar for communion. We take our angel David with us and give him some of the bread. However, he won't drink the grape juice. On this Sunday we were getting back into our pew when David had to sneeze. Well, the poor gentleman sitting in front of us was splattered. He had bread on his jacket and in his hair. Fred was picking bread out of his hair while the rest of the family was trying to keep from joining David in laughter. The poor man was very gracious, but has never sat in front of us again! We, and other church members, still have a laugh when we think of it!

Fred and Carolyn, angel David

What Angels Say...

- Here are the rules of the game... I throw it behind the couch and you go find it!
- Do you really need that full head of hair?
- Let's have another slumber party tonight!
- Surprise! I understand everything you say, so be careful!
- If I hear the sound of the microwave, I expect food!
- I am a lot smarter than you think, and I hide it sometimes to get out of WORK!
- When I laugh hysterically, it really, really IS funny!
- Someday I am going to hold you down, trim your fingernails, and see how YOU like it!
- A swimming pool =water= FUN TIME!
- Don't be in such a hurry to take me out of the bathtub!
- I see an iPad; I HAVE TO HAVE IT!
- If you take me out of my routine, I am really NOT going to be happy!
- Sometimes I just want to be a kid!
- Babies are OK... as long as I AM THE CENTER OF ATTENTION!
- I have to confess; sometimes it really is my fault instead of my sibling's!
- I'll take the macaroni and cheese!
- Wish you didn't have to work so hard taking care of me!
- Never give up on me!
- Sometimes you go a little overboard with the "I love you's"; but the truth is, I really love you a lot, too!

HYPOPIGMENTATION

Information from the 2009 document written by Charles A. Williams, M.D. and Sarika U. Peters, Ph.D.

[http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20as%202009%203-19-10.pdf)

When AS is caused by the large deletion, skin and eye hypopigmentation usually result. This occurs because there is a pigment gene (the P gene, also termed OCA2), located close to the AS gene, that is also missing [75]. This pigment gene produces a protein that is believed to be crucial in melanin synthesis. Melanin is the main pigment molecule in our skin. In some children with AS, this hypopigmentation can be so severe that a form of albinism is suspected [76]. When AS is caused by the other genetic mechanisms, this gene is not missing and thus normal skin and eye pigmentation is seen. Children with AS who have hypopigmentation are sun sensitive, so use of a protective sun screen is important. Not all AS children with deletions of the P gene are obviously hypopigmented, but may only have relatively lighter skin color than either parent.

Surveys of individuals with AS demonstrate an increased incidence of strabismus. This problem appears to be more common in children with hypopigmentation (as above), since pigment in the retina is crucial to normal development of the optic nerve pathways. Management of strabismus in AS is similar to that in other children: evaluation by an ophthalmologist, correction of any visual deficit, and where appropriate, patching and surgical adjustment of the extraocular muscles. The hypermotoric activities of some AS children will make wearing of patches or glasses difficult but many are able to accomplish this.

I

IEP

Michelle Harvey is on the ASF Family Resource Team. She has given presentations at the ASF biennial conferences. Go to: <https://www.angelman.org/resources-education/asf-family-resource-team/> to complete a form and contact her.

Individual Education Plan (IEP) Issues and ASF IEP Bank

**** To search the ASF IEP Bank or Add a Goal to the Team Bank:*

<https://www.angelman.org/resources-education/iep-bank/>

Instructions for adding an IEP goal to the IEP Bank:

<https://www.angelman.org/resources-education/iep-bank/>

About Michelle Harvey: As an ASF Resource Team member, Michelle's areas of focus are IEPs and ABA. If you have questions about IEP goals, IEP meetings with teachers, or your child's inclusion at school, Michelle is your go-to resource. Her MEd in Special Education (Severe/Comprehensive Disabilities) and ABA form a great foundation for her work with the ASF. She is also interested in the intersection of behavior/ABA principles and education and applying that to disabilities such as Angelman syndrome. Michelle was introduced to Angelman syndrome through her youngest brother (Matt, Del+), who recently graduated from high school and is now in his district's community-based program. She adores her brother and is very proud of him, crediting much of his success to her mom and dad. She enthusiastically uses her personal experiences to help other AS families. Michelle currently lives in San Diego.

ASF IEP Bank

Allows parents and IEP team members to exchange IEP goals and objectives with each other to assist in planning your student's IEP.

IEP Checklist iPhone App

The Parent Educational Advocacy Training Center (PEATC) developed this app to help parents of students with special needs become better-informed advocates by making IEP information easier to access.

Wright's Law

Find articles, law and regulations, and tips about how to get quality services in your child's IEP on this page. You'll also learn how to use tactics and strategies to negotiate with the school through IEP cases, recommended books, and free publications about IEPs.

Special Education Guide – IEP

Find a break down the IEP process and other special education issues. Find articles on the teacher's perspective, the parent's role in IEP meetings and tips on how teachers and parents can work together to create the best plan for the student.

Angelman Family Contributions: IEP

Take someone with you to the IEP meeting. One of you can write down main topics and comments and even jot down questions that come to mind while things are being discussed. The information can be overwhelming. Know your rights and go in knowing what you want, or with questions for your child's services.

Terry, angel Byron, age 35

Keep the goals simple and work on one at a time. Once one goal is gaining progress another can be added.

Donna, angel Douglas Joseph

Public school districts are not always funded adequately to provide the support services that each child needs; therefore, you must be an advocate for your child. Contact your state advocacy agency and ask them to come with you to IEP meetings. Take family and other parents of children with disabilities in your area to meetings for support. Never go alone to an IEP meeting because it can be a very emotional process and can often become overwhelming. When emotions are high you miss information that someone else could catch. The squeaky wheel gets the grease! Don't sign an IEP until the best interest of your child is met. When school districts say (and they will) that your child doesn't qualify for therapies, services, or special considerations; you need to remember that sometimes your child really does qualify. The school district just doesn't always have the means to make it happen.

Anne

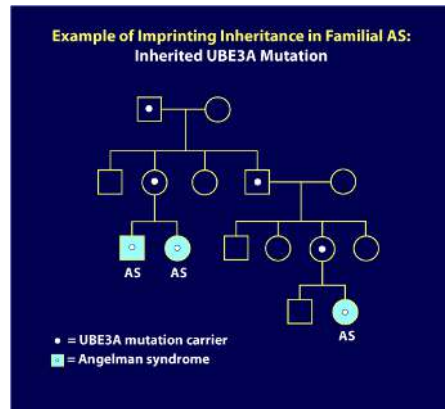
IMPRINTING DEFECT

(3% of cases) – These individuals may have a deletion of the imprinting center on Chromosome 15, but cases can also be caused by loss of imprinting information during the mother's oogenesis. Loss of imprinting will prevent expression of the maternal UBE3A gene in the brain. (50% recurrence risk)

IMPRINTING INHERITANCE

UBE3A mutations and Imprinting Center deletions can exhibit imprinting inheritance wherein a carrier father can pass on the genetic defect to his children without it causing any problems, but whenever a female passes this same genetic defect on to her children, regardless of the sex of her child, that child will have AS. The pedigree diagram below illustrates imprinting inheritance. Here, AS has only occurred after a carrier mother passed on the gene defect (for example as in the two siblings with AS pictured on the left lower part of the pedigree). In addition, a distant cousin in this family also has AS due to the imprinting inheritance. In the diagram, individuals with the light blue circles or squares have AS but everyone else in the family is clinically normal. The white dots represent asymptomatic, normal carriers of the AS mutation. When an AS genetic mechanism is determined to be inherited, genetic testing of

family members can usually identify carriers of the gene defect. As you might imagine, professional genetic counseling is advised in these situations.



INCIDENCE OF ANGELMAN SYNDROME

The answer to this question is not precisely known but we do have some estimates. A Swedish study of school age children showed an AS prevalence of about 1/12,000 (Steffenburg, S et al., 1996) and the Danish study of AS children in medical clinics, over an 8 year period when there was 45,000 births, showed a minimum prevalence of 1/10,000 (Petersen, MB et al., 1995). The most recent data comes from an excellent study from Denmark that used multiple, relatively complete ascertainment methods to determine a prevalence of 1/24,000 (Mertz, LG et al., 2013).

Several reports have tried to address the prevalence of AS among groups of individuals with established developmental delay. The results showed rates of 0% (Vercesi, AM et al., 1999), 1.3% (Aquino, NH et al., 2002), 1.4% (Jacobsen, J et al., 1998), and 4.8% (Buckley, RH et al., 1998). The Buckley paper extrapolated their data in order to compare it to the population of the state of Washington (using 1997 Census Bureau figures) and obtained an estimate of 1/20,000, a number similar to that often quoted, but not referenced in terms of methodology, in a 1992 review paper (Clayton-Smith, J and Pembrey, ME 1992).

There appears to be no reported prevalence studies that have screened newborns to detect rates of AS. Population wide prevalence figures would need to take into consideration that longevity in AS is probably reduced (severe mental delay and seizure presence would be risk factors) but no significant population-based data are available on life span shortening. Likewise, it is not known what percent of individuals with AS are undiagnosed, although this is expected it to be significant. Accordingly, to estimate the number of people with AS living in the society, it would be inaccurate to divide any estimated AS prevalence figure into a total population number.

In summary, AS is seen throughout the world, diagnosed in individuals of all races. Although the true incidence remains unknown due to challenges of early identification, misdiagnosis, etc., it appears that the prevalence of AS among children and young adults is somewhere between 1/12,000 and 1/24,000.

Charles Williams, MD

January 30, 2003; Reviewed 6-25-05 and Reviewed 11-4-08; Updated 10-8-2015

INCLUSION RESOURCES

See C- Community

<https://ahs.uic.edu/>

The Institute on Disability and Human Development

The IDHD is designed to give you updated information on public service, research, and academic programs and centers focused on removing barriers for people with disabilities and promoting their full participation in community living.

Parents' Perspectives on Inclusion and Schooling of Students with Angelman syndrome: Suggestions for Educators

Yona Leyser Ph. D.

Rea Kirk Ed. D Northern Illinois University, University of Wisconsin-Platteville 2011

<https://files.eric.ed.gov/fulltext/EJ937177.pdf>

Working with a Child who has Angelman syndrome / The Role of an Early Childhood Special Education Teacher in Kindergarten

Katherine M. Dobbs

St. Cloud State University, jkjrdobbs2@gmail.com 4-2017

http://repository.stcloudstate.edu/cgi/viewcontent.cgi?article=1019&context=cfs_etds

Angelman Family Contributions: Inclusion

We are very actively involved in our community. Our angel Maddy, age 12, loves going to the soccer field to watch her sister play, attending professional soccer games, and going to our school's plays, musical theater productions, or dance concerts. The more we take her out into the community, the more people get to meet her and are always happy to see her smile and having a great time wherever she goes.

Myriah, angel Madeline, age 12, Mutation

My angel Miata is 24 years old and very active in her community. We attend church, volunteer with local foundations, and are always on the go. Every weekend we are out and about, soaking up the sun and spreading joy and love to everyone we meet. We've even spoken with general managers at our local Walmart and was able to get them to order the Caroline Cart for easier shopping.

Patricia, lady729@yahoo.com, angel Miata, age 24

Any time we can, we include our daughter in our community events. She attends church, her brother's XC and track meets, and VBS. What we have discovered is that our daughter loves to go places, so she often has great behavior and lights up with the opportunity to be out and about with her family. On the other hand, some events are just not a blessing to her and end up being a ton of work and for something she could care less about. This is how we weed out her inclusion into community events. If I suspect she cares and wants to be a part, we make that our aim. Otherwise, we figure out a way for her to do something different or more on par with something she would enjoy. For instance, we used to have over 100 students at our house for special events. These nights simply overwhelmed her, so once we discovered this we tried to arrange something different for her, like going with a caregiver to a local store or going to our park. Another example is that for many years going to church was a huge struggle

but a high priority for our family. When the time was right and leadership at our church was favorable to the idea, I started a ministry to families with special needs. This has been such a blessing because not only does it help my daughter it expands to allow any family experiencing special needs to attend our church.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

Developing a community around your child is vital. Get involved at your church and other community groups.

AS Family Member

INTERNATIONAL RESOURCES

Save the Children

Save the Children is the leading independent organization creating real and lasting change for children in need in the United States and around the world. It is a member of the International Save the Children Alliance, comprising 27 national Save the Children organizations working in more than 100 countries to ensure the well-being of children. Save the Children responds to any emergency that puts at great risk the survival, protection, and well-being of significant numbers of children, where addressing the needs and well-being of those children is beyond the indigenous coping capacity, and where Save the Children is able to mobilize the financial and human resources to take urgent action on their behalf.

Catholic Relief Services (CRS – USCC)

Catholic Relief Services was founded in 1943 by the Catholic Bishops of the United States to assist the poor and disadvantaged outside the country. Working through local offices and an extensive network of partners, CRS operates on 5 continents and in over 90 countries. They aid the poor by first providing direct assistance where needed, then encouraging these people to help with their own development. Together, this fosters secure, productive, just communities that enable people to realize their potential.

United Nations Enable

United Nations Enable is the official website of the Secretariat for the Convention on the Rights of Persons with Disabilities (SCRPD) in the Department of Economic and Social Affairs (DESA) at the United Nations Secretariat. The website provides public information on topics related to disability and the work of the United Nations for persons with disabilities.

Association for the Advancement of Assistive Technology in Europe

The AAATE is the interdisciplinary Pan-European association devoted to all aspects of assistive technology.

INTERNATIONAL CONTACTS

Argentina: [Casa Angelman](http://CasaAngelman.org)

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Australia: [Angelman syndrome Association Australia](http://AngelmanSyndromeAssociationAustralia.org)

Email: wildkellie@gmail.com

Facebook: www.facebook.com/angelmanaustalia

Austria: [Angelman Verein Osterreich](#)

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Facebook: www.facebook.com/casscharity1993

Canada (Quebec): [Angelman Syndrome Foundation of Quebec](#)

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Facebook: www.facebook.com/Centre-de-r%C3%A9pit-Angelman-respite-center-Soci%C3%A9t%C3%A9-du-syndrome-dAngelman-162364910441918

Chile: [Familias Angelman Chile \(No National Association\)](#)

Email: www.facebook.com/angelmanchile/?ref=page_internal

China (Hong Kong): [Hong Kong Angelman Syndrome Foundation](#)

Email: info@hkasf.org

Facebook: www.facebook.com/HongKongAngelmanSyndromeFoundation/?hc_ref=SEARCH

Czech Republic (Slovakia): [Civic Association of Parents and Friends of Children with Angelman syndrome](#)

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Denmark: [Angelmanforeningen i Danmark](#)

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Italy: [Sindrome De Angelman – Italia \(No National Association\)](#)

Facebook: www.facebook.com/groups/17142459959

Japan: [Angel No Kai \(blog\) \(No National Association\)](#)

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Mexico: [Sindrome de Angelman Mexico \(No National Association\)](#)

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Netherlands: Angelman Syndroom Nederland

Email: info@angelmansyndroom.nl

Facebook: www.facebook.com/groups/141570629343929

Netherlands: Nina Foundation

Facebook: www.facebook.com/NinaFoundationNL

New Zealand: The Angelman Network and The Angelman Network

Email: Ursula – angelmannetwork@gmail.com

Facebook: www.facebook.com/groups/104686089609200; www.facebook.com/theangelmannetwork

Norway: Norsk Forening for Angelmans Syndrom

Facebook: www.facebook.com/Norsk-Forening-for-Angelmans-Syndrom-120579744727860

Philippines: Angelman syndrome Philippines (No National Association)

Facebook: www.facebook.com/AngelmanPh

Poland: No National Association

Facebook: www.facebook.com/razem.m.wiecej

Portugal: Associacao de Sindrome de Angelman de Portugal

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Facebook: www.facebook.com/AngelmanPT/?hc_ref=SEARCH&fref=nf

Portugal: Sindrome de Angelman – Portugal (No National Association)

Facebook: www.facebook.com/groups/angelman.portugal

South Africa: South African Angelman syndrome Group (No National Association)

Facebook: https://www.facebook.com/groups/1533193256919383/

Spain: Asociación Síndrome de Angelman

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Uruguay: National Association Angelman syndrome (No National Association)

Facebook: www.facebook.com/SINDROME-DE-ANGELMAN-EN-URUGUAY-148648096949

Venezuela: Angel Foundation in Venezuela (No National Association)

Facebook: www.facebook.com/Fundaci%C3%B3n-Un-Angel-en-Venezuela-104776379607999

Vietnam: Angelman syndrome Vietnam (No National Association)

Facebook: www.facebook.com/AngelmanVietnam

iPADS

Angelman Family Contributions: iPads

Make sure your entire family and social circle know how important iPads are to your Angel and ask to buy their old devices when they upgrade!

AS Family Member

Always, always, always have water proof iPad case on your iPad!!

AS Family Member

Have multiple iPads! One for communication, one for entertainment, and a backup one when they break either one!

AS Family Member

A great tool for communication apps and entertainment. The very thick foamy protectors are great! This is a great place to have personal photos and videos, and also to access kid-friendly videos and shows. Facetime with relatives is fun, too!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

J

JOGGERS

Amazon site:

https://www.amazon.com/s?k=special+needs+jogging+stroller&hvadid=78065451263412&hvbmt=be&hvdev=c&hvqmt=e&tag=mh0b-20&ref=pd_sl_3hcq3e8zyv_e

Adaptive Mall:

<https://www.adaptivemall.com/special-tomato-jogger.html>

K

KETOGENIC DIET

The Charlie Foundation <https://charlifoundation.org>

Provides resources and information about diet therapies including recipes, kitchen products and a worldwide list of locations where families can see a physician to be placed on the ketogenic diet.

Angelman Family Contributions: Ketogenic Diet

Our son started on this around age 3 when he was having a ton of atonic (drop) seizures and some grand mal seizures, and he stopped seizing and started independently walking within 2 weeks! We stayed on it for 4 years, and then were able to use only a small dose of seizure medicine since then. He's 18 now! We worked closely with our neurologist and a dietician to monitor things, and we prepared about 4 days of meals at a time into sectioned plastic containers as it required very specific measurements and ingredients of high fat and low carb items. If you can use a newer approach like LGI with similar effects, it's worth it!! We think our son was able to develop mentally and process information better when his seizures were controlled.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

My daughter has been on a modified keto diet for over two years. The tips that have helped me are to have a few items always on hand and to make sure I either bring these when we travel or pick them up when we arrive Here are her top 10 foods.: 1. Pepperoni (travels easily, yummy snack that needs no pairing); 2. MCT Oil (we can add this to pretty much anything); 3. Carrots (because she always wants a little something crunchy); 4. Peanut butter (for those before bed easy snacks); 5. Canned tuna or chicken with mayo (just in case there is nothing else for dinner she can eat); 6. Avocado (healthy fat with the bonus of dietary fiber to help with constipation); 7. Salad stuff (for the lunches she takes every day to school); 8. Eggs (she eats three for breakfast, and we usually keep boiled ones on hand); 9. Butter for cooking those morning eggs; 10. Fat Bombs (made with coconut oil, peanut butter, cocoa and stevia)

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

L

LIFE EXPECTANCY AND ADULT HEALTH

Young adults with AS continue to learn and are generally not expected to have deterioration in their mental abilities. Physical health in AS appears to be remarkably good. Although the severity or

frequency of seizures may improve with age there is likely to still be the need for some type of anticonvulsant medication. Mobility issues become a more predominant concern as the child with AS ages, often associated with concerns about obesity. Individuals with AS who have severe ataxia may lose their ability to walk if ambulation is not encouraged. Scoliosis can develop in adolescence and is especially a problem in those who are non-ambulatory [36, 49]. Scoliosis is treated with early bracing to prevent progression, and surgical correction or stabilization may be necessary for severe cases.

The main adult problems in those with AS are essentially continuation of problems present in childhood. These include problems of seizure control, difficult behaviors and abnormal physical movements. The movement problems cover areas of ataxia, diminished ambulation and scoliosis. Adults with AS, due to behavioral issues, are perhaps more likely to be given some type of neuroleptic medication, and side effects or sedative effects of these agents can be a health problem. The good news regarding adult health in AS is that many health matters are the same as those encountered in the normal population.

It does not appear that there is any major neurodegeneration event and that the brain per se (as determined by MRI) is able to withstand chronic seizure problems without obvious morphologic changes. There appears to be no apparent increased risk for malignancy or tumor problems and it appears that if longevity is diminished, it is only to a mild extent. Life span does not appear to be dramatically shortened in AS but may be decreased by 10-15 years. There are reports of AS individuals living beyond 70 years although there is as of yet no actuarial data that estimates life span [87, 88]. Hopefully, the results of a current study on natural history, funded by NIH, will provide important information in the near future.

LIZZIE SORDIA, Editor of *Angelman Today*

Lizzie was born and raised in Southern California. She married her high school sweetheart and moved to Florida, where they now live with their two boys Braden and Nathan. Nathan was diagnosed with Angelman syndrome shortly after his second birthday. After battling many seizures Lizzie decided it was best to leave her career in corporate America and end her privately owned family publication to care for her son. Nathan is now 11 years old and seizure-free thanks to the Low Glycemic Index Treatment. She has become very involved in the Angelman community by hosting the annual walk for the Angelman Syndrome Foundation in Orlando, doing fundraisers for the Foundation for Angelman syndrome Therapeutics, and participating with Weeber labs. Today, Nathan is a much happier and healthier boy, making improvements every day. She believes her son is not his diagnosis; he is so much more and so are all the other children with Angelman syndrome.

LONG-TERM PLANNING

See S- Special Needs Trusts

<http://specialchildren.about.com/od/longtermplanning/qt/fftrust.htm>

A special needs trust is a trust designed for beneficiaries who are disabled, either physically or mentally. It is written so the beneficiary can enjoy the use of property that is held in the trust for his or her benefit, while at the same time allowing the beneficiary to receive essential needs-based government benefits. There are administrative advantages of using a trust to hold and manage property intended for

the benefit of the beneficiary if the beneficiary lacks the legal capacity to handle his or her own financial affairs.

In the United States, such trusts provide advantages in helping beneficiaries qualify for health care coverage under state Medicaid programs, and also for monthly cash payments under the Supplemental Security Income (SSI) program operated by the Social Security Administration.

Angelman Family Contributions: Long-Term Planning

I cannot stress enough... Plan long term! I waited and then I was hit with a lot of decisions all at once. Plan... Plan... Plan!

Marsha

Long term planning is a necessary evil. It causes great sadness to imagine our angel's life after we are gone. But we must have a plan.

AS Family Member

LOW GLYCEMIC DIET

LGIT is a high-fat, limited-carbohydrate diet proven successful in reducing seizures in AS by up to 90%. Supervision by a physician familiar with the treatment is needed.

Angelman syndrome Clinic at Massachusetts General Hospital

Where a group led by Dr. Ron Thibert, D.O., neurologist and pediatrician at Massachusetts General Hospital, completed a clinical research study on LGIT.

Low glycemic index treatment for seizures in Angelman syndrome

Article published in Epilepsia about low glycemic index treatment (LGIT) about a study of six individuals on the LGIT diet and its findings.

The Charlie Foundation

Provides resources and information about diet therapies including recipes, kitchen products and a worldwide list of locations where families can see a physician to be placed on the ketogenic diet.

Angelman Family Contributions: Low Glycemic Diet

Do not kill yourself following a specific diet unless you see clear evidence in your child that it is beneficial!!

AS Family Member

M

MARATHON

Bank of America Chicago Marathon (2020) October 11

Windy City Angels: Angelman Syndrome Foundation

<https://www.angelman.org/event/bank-of-america-chicago-marathon-2020/>

The ASF has 27 guaranteed entry slots!

Or- contact Kitty Murphy kmurphy@angelman.org for 2020



and future marathons!

MARRIAGE AND PARTNERSHIPS

To avoid the “Who does more?” competition that breeds resentment and erodes marriages and relationships, it might be wise to get everything out in the open and create a list of responsibilities that involve caring for your angel. Plug in “who does what” on the list and come to an agreement. This plan will also help establish the all-important routines that help your angel thrive. Of course, there will need to be some flexibility at times, but, having a clear plan for the day-to day responsibilities helps lessen chaos and burnout. The pressures of having a special needs child can tip a marriage over the edge, so you HAVE to be proactive. Having a scheduled weekly respite “date”-doing anything together for the just the two of you- will help you keep connected. If one of the parents is not willing to do even a small share of the responsibilities, it is time to see a professional!

Sample list:

- Get angel up, go to the bathroom, and get dressed
- Fix breakfast
- Pack lunch
- Supervise breakfast and medications
- Brush teeth, comb hair, sunblock, etc.
- Wait for bus or drive to school or program
- Communicate with school and teacher
- Do laundry
- Do grocery shopping
- Do clothes shopping
- Monitor and order medication
- Doctor appointments
- Therapy appointments
- Prepare dinner
- Bathe
- Toileting
- Finger nail and hair trimming
- Bedtime routine (same time every evening as much as possible), tooth brushing, etc.
- Get up in the night, if necessary... maybe take turns?? :)

The following was written by: Drs. Charles and Elizabeth Schmitz. They are America's #1 love and marriage experts and the authors of the award-winning and bestselling book, *Building a Love that Lasts*. Life is not always fair, just and beautiful. And often life doesn't turn out as you had expected.

But the truth is, when you're dealt a "bad hand," you pick yourself up, dust yourself off and get back in the game. Giving up, feeling sorry for yourself, and crying over the unfairness of it all doesn't cut it in life. Parents of special needs children certainly know this is true.

Raising children with special needs challenges even the strongest marriage. But here is the truth: You cannot let your child's disability or ailment interfere or destroy your marital relationship. As we always tell our clients, based on our 30+ years of research around the world, "the parents' relationship with

each other trumps everything else!"

Here are 9 tips for nurturing your marriage while caring for a special needs child:

1. Talk openly and honestly about your feelings and frustrations. In times of stress, we often keep everything bottled up inside until it explodes out over the slightest disagreement. But this approach won't work if you want your marriage to survive and thrive. In successful marriages, there are no sacred cows. Happily married couples share insights about everything — the good, the bad and the ugly. They create a safe space let of steam and just listen to one another. They are each other's best friend.

2. Keep the flame of your love affair alive ... every day. Can you rattle off a list of activities, topics and destinations that light you and your spouse up and reconnect you to fun and romance? Have you figured out what clears your mind and gives you an unobstructed view of your world together? If not, start today by carving out time to have a romantic date with each other, get a hotel room, go for a long walk together, drink a bottle of wine watching the sunset, write each other a love note, or snuggle in bed a little longer in the morning. Little loving gestures made every day are what thread a successful marriage together.

3. Tackle all financial challenges with teamwork. Balancing the family budget requires teamwork, especially when the added challenge of taking care of a special needs child comes your way. It requires common goals. It most certainly requires family support. People in love support each other through thick and thin — through tough times and uncertainty. So lean in and figure out those finances together.

4. Don't blame each other when things get tough. The blame game never works in love and marriage, in fact it's destructive. It's easy in tough times to blame the one you love for your collective misfortune, but shaming or chastising each other in challenging times only makes things worse. Frustration is normal, but resist the urge to lash out at each other when pressure mounts.

5. Don't wallow in self-pity. It is a wasted emotion. No problem in all of history was ever solved by feeling sorry for yourself. Successful couples grab "the bull by the horns" and work for solutions. Pause to rest and comfort each other? Yes. But indulge in self-pity? No.

6. Prioritize time alone together. Yes, finding time alone is challenging, especially when your special needs child requires a great deal of constant care. But your relationship matters, too, and also requires constant care. Private time alone together is essential to a successful marriage. Connection and communication improve tremendously after refreshing your mind and spirit. You have to belong to yourself before you can belong to others.

7. Nurture a network family and friends. You and your spouse do not have to do this all by yourselves. Ask friends and relatives for help. Solicit support from aunts and uncles. Seek support from your neighbors. Don't be shy about asking for help. Don't be deluded into thinking you can do it alone (or should do it all alone). When you give concrete tasks or make specific, manageable requests, loved ones are usually happy to jump in and help out.

8. Recognize when you need professional care support. At some point, you may need help from professional caregivers, perhaps even an assisted-care or similar facility. Don't destroy yourself

with guilt if that time comes. At this critical juncture in your child's life, someone else may very well provide the best care. That is not a failing on your part. Make the choice that is healthiest for your whole family.

9. Tend to your own health and happiness. Engage in a daily exercise program. Sleep. Eat healthy — lots of fruits and vegetables. Take your vitamins! Make sure you both take your own medications on the prescribed schedule. And don't forget your annual physical exams. It is nearly impossible to take care of others when you don't take care of yourself.

Make no mistake about it, caring for a special needs child is sometimes stressful beyond belief. Don't let the negative impact shatter your marriage! The comfort and joy of a solid marriage helps you overcome almost any obstacles that comes your way.

MEAL TIME

ez pz Less Mess Happy Mats

It's a mat and placemat in one, which means that it captures all of your kids' messes. Plus, due to the unique design and silicone construction, the mat self-seals to the table, so no more tipped bowls or plates.

HipperBib

The Hipperbib doesn't look like a bib, it's a drool bib, is reversible, has secure snaps, & has a middle liner that won't let liquids soak through clothing.

Mum2Mum

We couldn't find a decent bib, so we designed our own – the super-absorbent Wonder Bib. It's the mess-proof, must-have for parents everywhere. You will love the quality, practicality and super-cool look of these bibs and accessories.

Angelman Family Contributions: Meal Time

Meal time can be mass chaos!! Our angel sits in an adult Rifton chair to prevent him from running around the kitchen seeking food!

AS Family Member

Make sure you have washable paint on your walls because your angel will throw food and you want to make sure it is easy to clean!

AS Family Member

The only bib that my three year- old son could not pull off on his own was a regular t-shirt.

Chaya, angel Avrumi, age 3.5, Del+

Pack an extra t-shirt in your angel's school backpack for the staff to put over your child's regular top at lunch time. When finished eating, the school can send home the dirty t-shirt in a plastic bag. This seems to be a more socially acceptable strategy for school.

AS Family Member

Take your angel out to eat, ask for a booth, let the waiter know she's a grabber! Take some food for her to eat while you wait for yours. Get her desert!!

Norma, angel Katarina, age 20, Del +

Our son did well using a spoon with sectioned containers so that he could scoop towards the side and keep the food on his spoon. He learned to use a fork similarly after that and it helps for plates to have a raised edge. We still use cups with lids and straws to prevent spills and promote independence.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

My son does not like to drink anything. It does not matter what it is. I had to come up with a way to increase his fluid intake. Dessert turned out to be the great motivator. I put the dessert on the table, just out of his reach and a glass of water in front of him. Then I tell him that he can have the dessert (or other desirable food) but he has to drink first. He empties that glass of water in no time at all!

Inge, ingediehl@hotmail.com, angel Marcus, age 34, Del+

MEDICATION

See A- Acid Reflux

See C- Constipation

See G- Gastrointestinal Issues

See S- Seizure Medication for detailed information and tables comparing seizure medications

<https://www.angelman.org/resources-education/resources/s/>

2009 Facts about Angelman syndrome

http://www.angelman.org/wp-content/uploads/2015/11/facts_about_as_2009_3-19-10.pdf

There is no agreement as to the optimal seizure medication although valproic acid (Depakote), topiramate (Topamax), lamotrigine (Lamictal), levetiracetam (Keppra), and clonazepam (Klonopin) are more commonly used in the North America. Carbamazepine (Tegretol), ethosuximide (Zarontin), phenytoin (Dilantin), phenobarbital, and ACTH are less commonly used. Vigabatrin (Sabril), an inhibitor of GABA metabolism, should not be used. [63] Single medication use is preferred but seizure breakthrough is common. Some children with uncontrollable seizures have been placed on a ketogenic diet, and this may be helpful in some cases. Children with AS are at risk for medication over--treatment because their movement abnormalities or attention deficits can be mistaken for seizures and because EEG abnormalities can persist even when seizures are controlled.

Angelman Family Contributions: Medication

Using saline nasal gel or spray helps for congestion.

Linda

To give cough medicine that tastes terrible, dip a bite of bread in the cough syrup and it soaks right up. What angel doesn't like bread?

Daniele Hill Smithfield, NC, angel Frank

We use Desitin cream not only for his bottom but also for sunburns. It works amazingly well.

Daniele, angel Frank

I purchased a pill container and every week I put all of his medications in there for each day. Then, I don't have to figure out or try to remember what he needs to take. It helps especially when you're in a

hurry.

Aina, angel Ra'Shawn

We use a product called Little Noses when our son suffers from nasal congestion.

Brisia, angel Mario

Melatonin has been a life saver for our family. Esmae takes 3mg dissolvable a half hour before bedtime and it generally allows her to sleep approximately 10 hours each night.

Julie, julialouis@howardhanna.com, angel Esmae, age 5, UPD

MOVIES/THEATER

<https://www.amctheatres.com/programs/sensory-friendly-films>

This is a link to AMC theaters which provides "Sensory Friendly Films" in partnership with the Autism Society.

Angelman Family Contributions: Movies/Theater

Justin loves movies! We probably have 150 of them! His favorites are old musicals: Singing in the Rain, Sound of Music, Oklahoma, Funny Lady, and Wizard of Oz. The new A Star is Born and Bohemian Rhapsody are top of his list, too!! He does like Disney movies too, but he does not like cartoons at all. They just do not hold his interest!

Barbara, bbarclay1@verizon, angel Justin, age 22

I will never forget the first time I took my daughter to a movie on Thanksgiving Day to see *Frozen*.

Extended family had come to visit, and I didn't want my girl left out, though I wondered if she would make it through the movie. She loved it! After this, we had so much fun as a family planning for another movie outing because this made us all feel so good that EVERYONE could do this. We discovered soon that she liked watching movies at home, too, and so because of her limited eyesight we mounted our TV above a sturdy piece of furniture, so that she can stand and watch her favorite movies and shows. I think this makes our girl feel validated and understood, but we could have so easily second-guessed our decision to take her to the movies, and we would have missed out on this thing she loves so much.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

MUSIC AND MUSIC THERAPY

Many Angelman syndrome patients are non-verbal and have difficulty communicating with parents or caregivers. To help these patients with their verbal, motor, and social skills, music can be used effectively in therapy.

What is music therapy?

In the established health profession of music therapy, therapists use music to address the physical, emotional, cognitive, and social needs of individuals. A music therapist assesses the strengths and needs of each patient and provides treatments such as singing, creating music, and listening or dancing to music.

The combination of music with therapeutic techniques can help strengthen patients' abilities, which can

promote improvement in other areas. Music therapy also provides a way of communicating for non-verbal patients.

How can music therapy help Angelman syndrome patients?

Tasks can be set to music to teach specific skills, such as teeth brushing while listening to a fun song, for example. Music can be used as a memory aid to teach specific information such as a phone number or address.

Music therapy can be used to help patients with walking by providing a steady rhythm for patients to improve their gait and stride. Therapists may play music with games to build movement skills. This may be combined with physical or occupational therapy to improve strength, flexibility, coordination, and range of motion.

Many patients with Angelman syndrome have a short attention span, which can limit social interaction. Music therapy provides a structured way for patients to interact with others. Through music, they can learn skills such as sharing, taking turns, and contributing to group activities. Specific songs can be used to build specific social skills, such as making eye contact.

How can I find a music therapist near me?

Your physician or physiotherapist may be able to recommend a certified music therapy center near you. Your physiotherapist may also be able to coordinate with the center to establish guidelines and goals for the therapy, as well as to track progress and address issues or concerns that might arise.

The following resources may be helpful:

Tuned in to Learning offers a comprehensive music-assisted learning curriculum for special education <http://www.tunedintolearning.com/>. American Music Therapy Association provides a listing of local music therapists. <https://www.musictherapy.org/about/find/>

Emily Malcolm, PhD

Angelman Family Contributions: Music

My son Brantley 2 (deletion +) loves listening to music! His Physical Therapist here recently is letting him stand with an Alltec baby boom box. You can adjust the volume. Brantley loves listening to the vibrations and rhythm! It's helping him wanting to move around more!

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

Our church provided a rhythm group for people with special needs and assistants so our son could make music with drums, triangles, sticks, bells, etc. He enjoyed having an activity of his own and a place to make a joyful noise! He's always enjoyed worship time in church, so this was a special treat.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

We play a CD in the hallway outside my son's bedroom since he enjoys falling asleep to music. This seems to help him stay put and calm down.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

N

Natural History Study

Natural History Study Overview

Participation in this study will help better our understanding of how development, behavior and communication change in individuals with AS over the course of their lives. We may know a lot about AS, but there are certain ages and issues that have never been studied. The AS Natural History Study will find out what life with AS is really like throughout the lifetime.

Participant Requirements

- Individuals with Angelman syndrome of any age are welcome.
- Participants will visit a study location once a year. At each visit, participants will have a physical exam as well as answer questions and complete surveys.

More Information

For more information and questions, call (617) 919-6008 or email ASNaturalHistoryStudy@childrens.harvard.edu.

The following are portions of a *Natural History Study* presentation given by Wen-Hann Tan at the 2019 ASF Conference:

Genotype-Phenotype Correlation in Behavior*

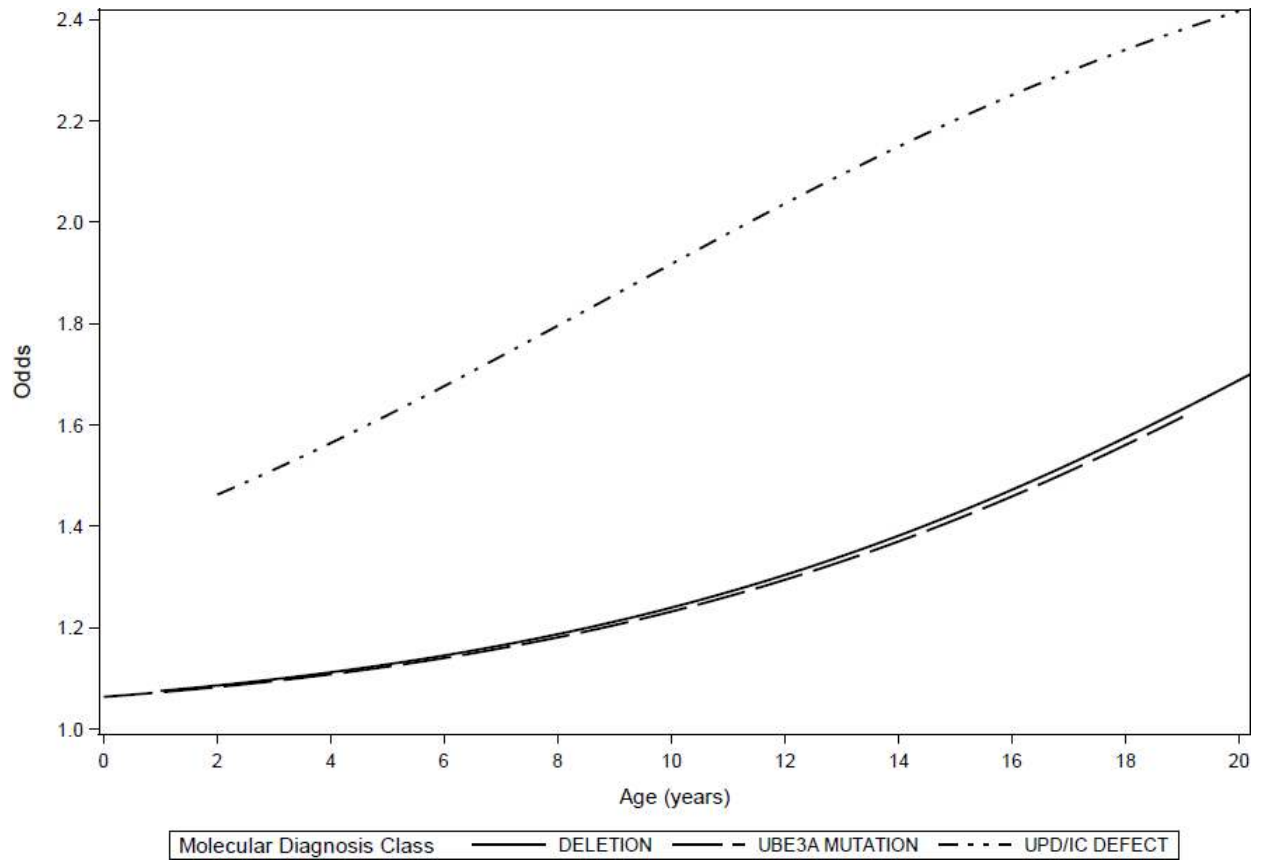
	Entire cohort (n=300)	Deletion (n=211)	UPD / Imprinting defect (n=56)	UBE3A (n=33)
Mouthing behavior	89%	92%	89%	70%
Easily excitable	88%	90%	91%	70%
Aggressive behavior	59%	51%	84%	70%
Anxiety	26%	19%	45%	36%

Temper tantrums	22%	17%	25%	52%
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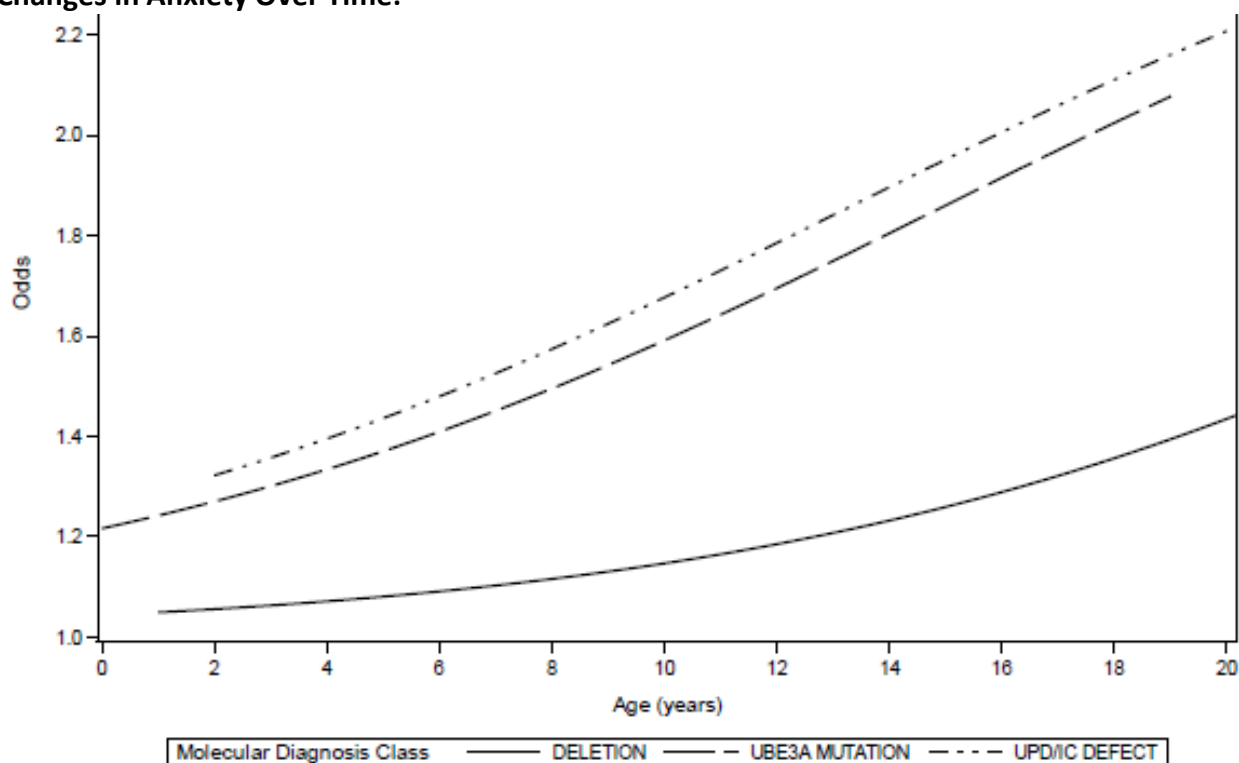
Well-known features less common than we think Subjects under the age of 5 in the first 2 years:

	Entire Cohort	Deletion	UPD / Imprinting defect	UBE3A mutation
Ataxic or broad-based gait	36/41 (88%)	21/22 (95%)	8/11 (73%)	7/8 (88%)
Clinical Seizures	60/92 (65%)	48/68 (71%)	6/13 (46%)	6/11 (55%)
Normal tone at evaluation	44/89 (49%)	24/65 (37%)	12/13 (92%)	8/11 (73%)
Easily-provoked laughter	55/91 (60%)	41/67 (61%)	8/13 (62%)	6/11 (55%)
Fascination with Water	69/92 (75%)	54/68 (79%)	8/13 (62%)	7/11 (64%)

Changes in Aggression (Pinching) Over Time:



Changes in Anxiety Over Time:



***Search PubMed by PMID number to read the studies and conclusions:

<https://www.ncbi.nlm.nih.gov/pubmed>

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NAUSEA AND VOMITING

BRAT diet

The BRAT diet is a diet for patients with gastrointestinal distress such as diarrhea, dyspepsia, and/or gastroenteritis. The BRAT diet is a bland diet that consists of foods that are low in fiber. Low-fiber foods were recommended as it was thought that foods high in fiber cause gas and possibly worsen gastrointestinal upset.

BRAT is an acronym for bananas, rice, applesauce, and toast, the staples of the diet. Extensions to the BRAT diet include BRATT (with tea), BRATTY (with tea and yogurt), and BRATCH (with chicken, often

boiled). Sometimes, the “T” represents tapioca. In addition to dietary restrictions, medical professionals recommended that all patients, regardless of age, drink plenty of fluids to prevent dehydration, along with oral rehydration solutions to replace the depleted electrolytes to avoid salt imbalance. Severe, untreated salt imbalance can result in “extreme weakness, confusion, coma, or death.

Foods to avoid if you suffer from nausea include:

- Greasy, oily, and spicy foods
- Milk and dairy products
- Alcohol
- Caffeine
- Raw vegetables like corn, cabbage, onions and beets
- Salmon and sardines
- Pork and veal
- Fruits such as oranges, grapefruits, cherries, berries, and figs

If you or anyone you know is suffering from any of the following symptoms along with nausea, contact your doctor or emergency room immediately:

- If you have been vomiting for more than a day with no respite
- If there is no retention of fluids for 8 hours if you are a child or 12 hours in the case of adults
- If there is blood or a coffee-ground like substance in your vomit
- If you have a headache, stomachache or stiff neck
- If you are exhibiting signs of dehydration such as dry mouth, sunken eyes, increase in thirst, and infrequent urination
- If your child has taken aspirin
- If your child is lethargic or unusually irritable
- If the nausea is prolonged
- If home remedies are not working
- If there was an head injury
- If there is a fever (especially in the case of children)
- If there is a feeling of confusion and lack of alertness
- If there is a rapid pulse or irregular heartbeat
- If there is difficulty in breathing

Angelman Family Contributions: Nausea

Before our daughter has any surgeries with general anesthesia, we request the medication Zofran to prevent nausea!

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del+ Class 1

O

OCCUPATIONAL THERAPY

If your child is going to see an Occupational Therapist they may be looking at the following:

1. **Self-Care:** being able to feed themselves with a fork and spoon, being able to drink from a cup and straw, being able to use their tongue, lips and cheeks to move food and chew, and being able to assist with dressing activities.
2. **Fine motor/visual motor activities:** how they use their hands to grasp small objects, how they hold crayons, how they play with toys and their skill level with their hands.
3. **Sensory:** how they respond to sensory experiences outside and within their bodies. Depending on the age of your child, their functional level, their needs and abilities will determine activities to work on at home.

Good activities in general for children:

- Play at the park: playing in the sand using their hands to dig, find toys in the sand to put into a container, digging with a shovel to put sand into a bucket and dump into a larger bucket. Supervision is important to make sure that sand and objects do not go into the mouth. Going on slides and swings with assistance as needed. Movement is important part of learning how our bodies work and helping us develop our balance reactions.
- Practice picking up different sizes and weights of objects and putting them into various sizes of containers, making sure objects do not go into the mouth.
- When taking a bath try painting on the inside of the bathtub with shaving cream, working on making lines that go down, across, and circles. When finished try squeezing a small easy to use squirt bottle with water to erase the paint (squeezing the bottle will help work on small hand muscles).
- Outdoors take large sidewalk chalk and practice drawing, scribbling, making vertical, horizontal lines and circles on the driveway or sidewalk. Erase it by using a squirt bottle to squirt water on it or try taking a sponge in a small bucket of water, squeeze the water into the bucket or on top of the drawing.
- Practice closing lips on various sizes of whistles to work on blowing. Try blowing a bubble off a wand that has been caught for them.
- Practice drinking from a straw using box drinks with a straw that the liquid can be gently squeezed up the straw working on them trying to suck the liquid up the straw themselves. Once they can drink from a straw practice using oral muscles by drinking thicken liquids from a straw (you can make your own smoothies using applesauce with juice, or yogurt with milk, or pudding with milk).
- Practice putting on shoes using your shoes or bigger shoes which may be easier to slip on.

Angelman Family Contributions: Occupational Therapy

Have fun trying activities with your child, remembering that children's work is play and that is how they learn.

Debbie Cahill, Occupational Therapist, Rady Children's Hospital, San Diego, CA

Learn and follow through with what the therapist is trying to accomplish. Let the therapist know what goals you and your angel are working on, and ask for her to teach you what to do at home. AND, if you

feel you are getting burned out doing all these therapies, TAKE A BREAK from them. You can't do it all!
AS Family Member

ORTHOTICS

Orthopedic Footwear

<https://ablegaitor.com/>

Angelman Family Contributions: Orthotics

The best shoes to use with orthotics are from the company *Billy Shoes*. The company is super helpful and accommodating.

Chaya, angel Avrumi, age 3.5, Del+

If one needs to protect a pic line or IV site in an adult Angel's arm area, use small-size knee immobilizers on your angel's arms. Tie (with an ace wrap) the top of immobilizers together in the back of shoulder area to keep them on the arms. It limits your angel's access to the IV or pic line site and makes it harder for them to be removed.

Karen, angel age 27, Del+

Our son started out having his tight heel cords snipped and receiving Botox injections to each calf before getting casted for ankle braces and then eventually using simple shoe inserts. This all seemed to help straighten out his feet, legs and his walking.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

P

PARENTS' HEALTH

The Burnout that Special Needs Parents Experience

Heather McCain

https://www.mombieneedscoffee.com/the-burnout-that-special-needs-parents-experience.html?fbclid=IwAR1fZtkfVrJBWVbXkdq_WA_WGndV5jSY1uiMoQs_uMRirolY9vHNemeliaY

I think most parents probably feel burnt out at some point while juggling all of the responsibilities that parenthood brings. It seems there is always a never-ending list of things to do, but never quite enough time to get it all done. Parents of special needs children have lists that are a bit longer, but still have only 24 hours in a day.

Our lists include medication administration, regular appointments, multiple forms of therapy, paperwork, unending phone calls, IEP meetings, learning to use and then using medical equipment, and so on. We often need to feed, change, or bathe our children who are well beyond their pre-school years. We need to make sure we don't run out of the medications that our child's life depends on, or diapers in a size that can't be bought in a store and must be purchased from medical supply companies. When we plan outings, we must make sure our destination will be accessible for our child. If you can imagine, I've only just put a dent in all the extra things a special needs parent must do, remember, or know.

Before anyone gets the idea that I'm complaining, I want to make sure to be clear that that isn't what this is about. I'm just sharing experiences. I understand that it can be hard for those who have not lived

this life to grasp what our day-to-day routine consists of (I use the word “routine” loosely) so I wanted to try to paint a picture as I’m getting into what this is all about, which is the burnout that special needs parents experience. This is something that I personally experience, and that I hear or see other special needs parents talk about often.

I’m not talking about that exhaustion at the end of a long day.

I don’t mean the needing a cup of coffee or two, or three kind of exhaustion.

I mean that deep down, all the way to your core exhaustion that creeps into your heart and mind, the kind that’s already there when you wake up in the morning.

It’s when you’re so burnt out that you can’t even bring yourself to open a piece of mail or check your voicemail because you can’t fathom adding one more thing to your list, not even a seemingly tiny task like returning a phone call.

It’s the kind of exhaustion you feel as you wake up to change a diaper and bed sheet in the middle of the night, like you have for the past 9 years. Or 20. Or 42.

It’s letting go of careers and plans.

It’s feeling like you have little control over what happens to your child, when you desperately want to protect them.

It’s handling meltdowns like a pro in public and hiding in the bathroom to cry later.

It’s watching monitors until the sun rises, even though you’ve been awake for approximately 29 hours already.

It’s waking up at 4 a.m. to make it to your child’s appointment at the specialty doctor 5 hours away.

It’s the heavy guilt of being away from your other children as you sit in the hospital with one, weighing on you like a boulder.

It’s being afraid of the future but learning to live in the moment—Who will take care of my child if something happens to me? What if my child outlives me? What if I outlive my child? Breathe. My child is here with me now. Enjoy this moment.

It’s falling into bed thinking, “how can I keep doing this every day?” Then, getting up the next morning to do it again.

It’s when our health suffers, mentally and physically. Depression and anxiety are common in parents of special needs children, and lifting a growing child and equipment such as wheelchairs takes a toll on one’s body over the years.

The burnout is real. I’m not telling you about it for pity, I’m simply sharing the reality of many with you. And it has nothing to do with how much we love our children. Trust me, we love them so much that we put their every need above our own. We love them so much that we wouldn’t trade being their parent

for the world. Know what else? You will probably never recognize how burnt out we really are on the inside when you see us with our children. That's because you will see us playing peek-a-boo, or beaming with pride over them, or kissing their soft cheeks over and over just soaking in all the love they offer. You hear the praises we give them and how we gush over every little thing they do. The burnout is real, but so is the unconditional love between us and our children.

You see us in the moments that keep us going, the moments that make all of the hard parts worth every second. But rarely does anyone see us at our most vulnerable, so rarely does anyone notice how burnt out we are.

The next time you see a parent of a special needs child, instead of saying, "I don't know how you do it" (trust me, we're not even quite sure how we do it), or "I couldn't do what you do" (trust me again, you could if it were your child), consider smiling and telling us we are doing a good job. Sometimes, that's all we really need to hear to keep on keeping on.

Angelman Family Contributions: Parents' Health

Parents often "turn off" their needs to ensure they're fully providing for their child(ren). I encourage parents to take moments throughout the day to recharge. We parents cannot function well, or be our best, if we aren't mentally, physically or spiritually healthy.

Annie, angel Ava, age 7, Del+

PEDIATRICIANS

Reference Guide for Pediatricians: *What Every Practitioner Needs to Know*

Susan Debrosse, 2017

<https://www.cancertherapyadvisor.com/home/decision-support-in-medicine/pediatrics/angelman-syndrome/>

PHOTO BOOKS AND PHOTO BLANKETS

Personalized Photo Blankets:

https://www.collage.com/photo-blankets?lc=MyBlanket&utm_source=bing&utm_medium=cpc&utm_campaign=BlanketVouchersExactMatch&msclkid=c632ae56922b1f646d7ff39768ee761e

Angelman Family Contributions: Photo Books and Blankets

Our 35 year-old son is fascinated with photo books. He will look at each one carefully. He smiles when he recognizes people. He is not interested in other books.

Susan, AS Mom

It is easy (and inexpensive) to create personalized photo books through the Costco photo website and many other online sites, too. I always choose the hard cover option. The books are great for birthday and holiday gifts! You can add humorous captions and read through the books with your angel! While reading, you can also say, "Point to the ..."

Alice, sandiegoasfwalk@gmail.com, angel Whitney, age 38, Del+

Our angel loves looking through pictures so we have a lot of durable photo albums or photo books for him to look through. Displaying photos in a clear wall hanging with picture slots is another great option. *Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1*

PHYSICAL THERAPY

Therapies 4 Kids

Therapies 4 Kids is an intensive therapy program for children with neurological disorders who have physical disabilities and are in need of Pediatric Intensive Therapy.

Angelman Family Contribution: Physical Therapy

My son Brantley 2 (deletion positive) is working with a PT from First Steps. I highly recommend early intervention with therapy services for any AS child! He has already come such a long way, to get to where he is now. Also it's great to get a therapist that pushes your child to keep going!

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

I want to share something that our physical therapist told us that might be helpful to other Angelman families. During a session one day, I remarked that my son Jack was babbling more during PT than he does during his speech therapy sessions and she told us that movement encourages speech! Who knew! So if it possible to incorporate some sort of movement during speech therapy (rather than just sitting in a chair) that might encourage more babbling. I definitely find this to be true for our son Jack!

Sayoko, sayoko.murase@gmail.com, angel Jack, age 2, Del+

POTTY TRAINING

See T- Toileting

PRODUCTS

Contact Lizzie Sordia, who is a member of the ASF Family Resource Team.

<https://www.angelman.org/resources-education/asf-family-resource-team/>

General Resources (Supplies, Durable Medical Goods, Products, Daily Living Products and Services).

Lizzie is mom to two young boys, Braden and Nathan (Del +). She joins the ASF Resource Team specializing in General Resources (supplies, durable medical goods, products, helpful daily living products and services). From finding the most appropriate beds for your child to giving you helpful tips to get you through a day as an AS family, Lizzie is the gal to contact. Lizzie is also creator of the online magazine Angelman Today and is passionate about raising awareness about AS and about the health of her children. Lizzie is the also the site coordinator of the Orlando, Florida ASF walk site!

Angelman Family Contributions: Products

Secura is a product by Smith & Nephew. It is a "total body foam cleanser" and is excellent at getting rid of odors, such as urine and feces.

<http://www.smith-nephew.com/key-products/advanced-wound-management/other-wound-care-products/secura/>

Mindy, angel Daniel, Del +

This is a good “recipe” for stain removal.

1 tsp. Dawn

3-4 T. hydrogen peroxide

2 T. baking soda

Scrub on with a scrubbing brush.

Simply Thick is a food and beverage “thickener gel product” for individuals who have swallowing difficulties. Daniel eats everything now since I started using the product.

www.simplythick.com

Mindy, angel Daniel, Del +

Q

QUESTIONS?

Contact the ASF:

<https://www.angelman.org/about/contact-asf/>

Email: Complete form at this link or email info@angelman.org

Angelman Syndrome Foundation

75 Executive Drive, Suite 327

Aurora, IL 60504

Phone: (630) 978-4245 | 800-432-6435

Fax: (630) 978-7408

ASF Family Resource Team: Complete form at this link to email.

<https://www.angelman.org/resources-education/asf-family-resource-team/>

QUOTES

Life is 10% what happens to you and 90% how you react to it.

On particularly rough days when I’m sure I can’t possibly endure, I like to remind myself that my track record for getting through bad days so far is 100% – so that’s pretty good.

If the purpose of life is to love deeply, live selflessly, and give generously, then I would say that the lives of special needs parents are among the greatest and most fulfilled. They are the true unsung heroes of the world.

I thought I would have to teach my child about the world. It turns out I have to teach the world about my child.

Parents who have children with special needs, also have special needs. They need to know more than the average parent. They need to do more than the average parent. They need more patience than the average parent, and so much more.

Life isn’t about waiting for the storm to pass...it’s learning to dance in the rain.

Believe in yourself as a parent. You are your child's best therapist and advocate.

Once you learn to appreciate small victories, there is no need for a finish line.

With tremendous burdens often come enormous gifts. The trick is to identify the gifts, and glory in them.

We must be willing to let go of the life we have planned, so as to have the life that is waiting for us.

Everybody is a genius. But if you judge a fish by its ability to climb a tree, it will live its whole life believing it is inadequate.

Being happy doesn't mean that everything is perfect. It means you've decided to see past the imperfections.

Accept what is, let go what was, believe in what will be.

Normal is an illusion. What is normal for the spider is chaos for the fly.

I've never met a strong person with an easy past.

Once you accept that your child will be different, not better or worse – just different – that's the first step.

None of the tests, assessments, rating scales or progress reports will ever be able to measure just how awesome my special needs child is or how much joy, happiness, and love he gives to the world.

The breaking of that awful feeling of isolation is the greatest gift one Angelman family can give to another.... Those of you who will share your experiences will do it because in the Angelman family, that is what we do. Audrey Angelman

R

RECURRENCE RISKS

See G-Genetic Counseling

RESEARCH: Current ASF

<https://www.angelman.org/research/>

Also, visit the **ASF Funded Research** 1996-Present

<https://www.angelman.org/research/asf-funded-research/>



The Angelman Syndrome Foundation (ASF) has invested more than \$10 million in Angelman syndrome research to date, supporting projects worldwide in the quest to find treatments for Angelman syndrome and ultimately a cure. Treatments resulting from the ASF's \$9 million investment in research help individuals with Angelman syndrome live better lives today and lead to better lives tomorrow, but

require ongoing financial support.

The Angelman Syndrome Foundation is dedicated to funding the highest caliber of research on Angelman syndrome. It is our hope that these funded researchers, and their collaborators and peers, will bring forth new discoveries that ultimately lead to safe and effective therapies that improve the quality of life for all with Angelman syndrome.

Active Angelman Syndrome Research Studies 2019:

<https://www.angelman.org/research/participate-in-research-studies/>

There are Angelman syndrome research studies happening at universities and medical centers throughout the country. You can help Be the Cure and advance research by having your family or your individual with Angelman syndrome participate in research studies. Studies may involve a phone interview, surveys or mailing of information. Some studies may require you to travel to other cities. Others may involve blood tests, EEG's and other medical procedures. See the list of **(2020) active studies below** to find out if there is a study that is right for you and your family.

As new studies become available the ASF will contact known AS families with the opportunity to participate. Make sure to complete the contact registry to stay informed.

Contact Registry: <https://www.angelman.org/contactregistry/>

FRESIAS Endpoint Study

<https://www.angelman.org/studies/freesias/>

Roche is sponsoring a non-drug study that will help to prepare for future clinical drug trials in AS. The data obtained will serve as a reference point or baseline for future drug trials. Current participating clinics are in Houston, Chapel Hill and Chicago.

Vanderbilt Music Survey

<https://www.angelman.org/studies/music-study-at-vanderbilt/>

An online, 30 minute survey for families of children 6 months to 5 years old. The study will ask about musical experiences and social and emotional well-being.

Phase 3 NEPTUNE

<https://www.angelman.org/studies/neptune-phase-3-clinical-study/>

The NEPTUNE study will evaluate the safety and effectiveness of OV101 on various aspects associated with Angelman syndrome in individuals aged 2 – 12 years old.

Early Development in Angelman syndrome <https://www.angelman.org/studies/panda/>

Conducted through Purdue University, this telehealth study aims to improve understanding of how children with Angelman syndrome develop in early childhood. Participants must be 18 months or younger.

Experiences of Adult Siblings

<https://www.angelman.org/studies/experiences-of-adult-siblings-study/>

Conducted through Texas Woman's University, this survey study aims to understand the experiences of adult siblings of individuals with intellectual disabilities.

Brain Imaging Study

<https://www.angelman.org/studies/brain-imaging-study/>

The University of North Carolina (UNC) School of Medicine is now recruiting participants that are Deletion Positive up to 5 years old, to examine the brain development of children with Angelman syndrome.

Angelman syndrome Natural History Study

<https://www.angelman.org/studies/angelman-syndrome-natural-history-study/>

This study at Boston Children's and the AS Clinic at Rady Children's Hospital, San Diego will help better our understanding of how development, behavior and communication change in individuals with AS over the course of their lives.

Research Interview

<https://www.angelman.org/research/participate-in-research-studies/research-interview-about-as/>

A non-medication research interview study seeking information about symptoms and effects on daily life that children and adults with Angelman syndrome experience, from the caregiver's perspective. Participants agree to a 60-90 minute telephone interview.

Caregivers of Children, Adolescents & Adults with AS

<https://www.angelman.org/studies/research-interview-about-as/>

This is an interview study to gather information about caregivers' experiences with Angelman syndrome (AS) to better understand the symptoms and impacts of AS. The study is sponsored by Ovid Therapeutics, and conducted by ICON, a health research organization.

Augmentative and Alternative Communication (AAC) Immersion Project

<https://www.angelman.org/research/participate-in-research-studies/augmentative-and-alternative-communication-immersion/>

This study at Portland State University will evaluate the impact of communication partner coaching in Augmentative and Alternative Communication (AAC) intervention.

Extraction of Neuronal Stem Cells from Dental Pulp for Human Neurogenetic Disease Studies

<https://www.angelman.org/research/participate-in-research-studies/extraction-of-neuronal-stem-cells-from-dental-pulp-for-human-neurogenetic-disease-studies/>

Dr. Lawrence T. Reiter at the University Of Tennessee Health Science Center in Memphis, TN is conducting a research study to determine if neurons can be grown from the dental pulp of individuals with various neurogenetic syndromes including chromosomal duplications and deletions of human chromosome 15q.

Harvard Music Study

<https://www.angelman.org/research/participate-in-research-studies/harvard-music-study/>

Researchers at Harvard are studying how people with AS respond to and participate in musical activities, and how parents interact musically with their children. In the study, the researchers will play your child a series of recorded songs and poems while recording his/her physiological activity. They will also ask parents to complete surveys about your experiences as a parent and about your child's interests in music and other arts activities.

Longitudinal study of how children with AS develop over time

<https://www.angelman.org/studies/longitudinal-study-of-now-children-with-as-develop-over-time/>

The Purdue Neurodevelopmental Family Lab at Purdue University is recruiting participants nationwide for a research study on the early development of children with Angelman syndrome. Families of children ages birth to 48 months complete online forms and optional phone interviews annually for 3+ years. The surveys and interviews cover topics about general child development (e.g. thinking, motor, social skills), medical needs, problem behaviors and child strengths.

RESEARCHERS

Please visit the ASF website for the Researcher names and affiliations.

<https://www.angelman.org/resources-education/resources/r/>

Also, visit the *ASF Funded Research 1996-Present*

<https://www.angelman.org/research/asf-funded-research/>

RESIDENTIAL PLACEMENT

Angelman Family Contributions: Residential Placement

We kept our daughter at home until she was about twenty-one years of age. Deciding on group home placement was the most difficult decision we ever made. At that age she was not sleeping at night and some of her behaviors had gotten worse. Around that time we read an article written by a doctor who had a special needs child. In the article he explained that after his wife's death, he had a very hard time caring for their son. His son was now up in his forties and the doctor was close to seventy. He said it was a very hard adjustment for both of them after his wife passed away. He then mentioned something that helped us make the decision with a better conscience. He stated that he wished he and his wife had been able to ease their son into residential placement sooner because their son would have gotten to know the program better and vice versa. He went on to write that he and his wife would have known they weren't abandoning their son because they would still be there to monitor it all and help him to adapt. They would still see their son as often as they wanted. His reflections helped us in this process and now our daughter has been in a group home now for about ten years. She has her own room with other housemates. We pick her up weekly for outings, if not more. We can say with confidence that it was the best decision we could make for her as well as for us as we all grow older

Linda

When Tip was thirty years old we had an opportunity to place him with a residential program and we took advantage of that. Though he is two hours from home, we are able to bring him to our/his house whenever we want. It is still a challenge to take care of him, but he moves about the house as if he's never left. We feel blessed to be able to bring him home, cut his hair and sit with him on the porch. In

the last twelve years we have learned how best to advocate for him and we are still learning. This was the right decision for us.

Jane, angel Tipton,

Thoughts of the future can cause a great deal of stress for parents, siblings, and other family members of individuals with Angelman syndrome. Fears about where the child will live, who will care for him or her, and quality-of-life issues are common. It is never too early to think about and begin planning for the time when the angel might move out of the home. This time is different for all families and each family must choose what is right for them. It cannot be stressed enough that the transition process from the angel's home to residential placement, no matter what the age of the person, should involve the entire family. Encourage questions from siblings, grandparents, and other family members involved with the angel. Take siblings to visit the new residence so that they will know where their brother or sister will be living.

AS Family Member

RESOURCES

ASF Family Resource Team: <https://www.angelman.org/resources-education/asf-family-resource-team/>

***A form is provided for you to contact Michelle, Lizzie, and Dr. Eric.

Michelle Harvey: Individual Education Plan (IEP) Issues, ASF IEP Bank, Educational Issues and Advocacy.

Lizzie Sordia: General Resources (Supplies, Durable Medical Goods, Products, Daily Living Products and Services).

Dr. Eric Wright: Resources and Services specific to State Waivers, Government Assistance, Insurance and Advocacy.

Champions

See C- Champions for a list of Angelman syndrome parents who you can contact for specific information and support.

Educational Programs and Resources: See the ASF website at: <https://www.angelman.org/resources-education/>

***Specific information about many of the resources is featured throughout *A to Z!*

Education

- [Communication Book by Dr. Calculator](#)
- [Communication Training Series](#)
- [Educational Webinars](#)
- [IEP Bank](#)

Resources

- [ASF Family Fund](#)
- [ASF Family Resource Team](#)

- [International AS Resources](#)
- [Federal Resources](#)
- [State Resources](#)
- [Regional Representatives](#)
- [Professional References](#)
- [AAC & Technology](#)
- [Education Resources](#)
- [Employment Resources](#)
- [Financial Planning](#)
- [Household Resources](#)
- [Inclusion Resources](#)
- [Recreational Resources](#)
- [Seizure Resources](#)
- [Special Needs Safety](#)
- [Support Resources](#)
- [Therapy Resources](#)
- [Travel Resources](#)

Angelman Family Contributions: Resources

The National Association of Child Development has been our family's most important resource for many years. We deeply appreciate NACD's invaluable perspectives about what we can do at home to help Carly grow to her potential, whatever that may be. They have helped us with sensory issues, mobility, nutrition, seizures, sleep, and oral motor and communication needs. They are our main go-to when we have questions or want to be prepared for doctor appointments. They have helped us understand how to view Carly's challenges holistically, considering the interplay between many factors. They help us address underlying issues rather than just treat symptoms. They help us maintain focus and healthy balance in our home and they show great respect for our family's goals and values. Investing in Carly's health and well-being has taken much physical, mental and emotional energy. I want the best for all of my children. Sometimes we have had to be persistent and patient for long years before recognizing the full reward of those efforts. Tremendous blessings continue to unfold and we're so grateful!

<http://www.nacd.org>

Lisa, angel Carly, age 16

RESPITE WORKERS

Angelman Family Contributions: Respite Workers

Treat them well, offer plenty of training, and pray that they will stay.

AS Family Member

Always go through and consider wisely when making a decision on selecting one. Take the time to research, interview, and ask around!

Sarah, j.sorrels09@blueriver.net, angel Brantley, age 2, Del+

Realize that respite workers will come and go, especially as your child becomes an adult.

AS Family Member

Find your own staff whenever possible.

AS Family Member

Our child's respite workers are his friends!! They do wonderful things with him that we are too tired to do. They have taken him to fraternity parties, amusement parks, concerts etc. The more fun they have together the better they get along.

AS Family Member

Trust that your angel will let you know if they don't like a caregiver! Ours will hit and throw things at certain respite workers that he does not like.

AS Family Member

Let your staff know that you have cameras throughout your house when you first hire them. If they have a problem with that, they can take a hike!

AS Family Member

Trust your gut! If you think it isn't going well with a respite provider, do not ignore that feeling. Moms and dads are always right!

AS Family Member

Social workers where your kids receive therapy can be a great source for how to find respite help and even financial support for them. Many of our helpers have been college students at a nearby school in majors related to special education or sports therapy.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

ROUTINES

Angelman Family Contributions: Routines

Find activities in the community your child enjoys. Develop a routine to take your child into the community. For example, go daily or weekly to a park, or go on short visits to a store or a recreation center. Most public places will try to accommodate your angel if they know your expectations.

Donna, angel Douglas, age 25

Begin any kind of daily living routine early and keep practicing. It may take years before your child can do the skill independently, but it is worth it. This includes hand over hand use of a spoon, pulling pants up and down when toileting, and bathing. I began putting deodorant on my child long before she really needed it so that she would be used to the sensation by the time it was truly important.

Laura and Jess, angel Leah

Establishing a routine early on has helped calm my son Coby. We dim the lights and turn down the television at about the same time every night. We then put him into bed at almost the same time and lay with him until he falls asleep. His older siblings also know to use gentle voices at night.

Maria

We try to keep to the same routine at home as much as possible and this helps with language understanding. We can say "It's time for a bath!" and our daughter knows what that means. For eating, our daughter finally has the hang of using a fork. We tried for a long time with a child fork, and then we realized a sharper adult fork actually made it easier for her to stab the food. Our occupational

therapist experimented with a bowl that was held at an angle so that our daughter could stab the food more easily.

Christine and Giovanni, angel Chiara

Routine is essential for our Angelman child. It assists with normalizing his wake/sleep cycle and keeps his behaviors to a minimum if he knows what to expect as the day progresses.

Terri

We are big on routines. It worked for our typical child, so we continued with it. We are not sure if we are just lucky or if it is our routines that help our angel have a pretty normal sleep pattern.

Leah, angel Emily, Del +

I believe in being consistent with how to react to things, but I don't believe in routines for mealtime or bedtime. The reality in life is that you are not always able to stick to a routine.

Carol and Rick, angel Rob

My angel loves to help out with chores around the house. I usually give her a dust rag or paper towel and let her help me with chores. She loves it and I get a lot done!

Steve and Lee Ann, angel Ella, UPD

Routine. Routine. Routine. I have found that if Kade stays on a routine, all other areas improve.

Hollie

Routine, routine, routine! My son benefits greatly from having a routine!

Danielle and John, angel Alex

Routines are very important with my little angel. Having her get used to a specific routine is hard from the start, but when she is used to, she won't go back. My 3 year old angel likes to stand while eating, but we taught her to sit before she eats. Now, she never eats or even drinks water unless she is sitting on a chair.

Angel aged 3

We feel that a very important part of routines is something the Angelman person can do to contribute to the household. Our daughter is responsible for putting her adult pull-ups away in her closet every month when they are delivered. She isn't always interested in initiating the activity, but once she has put each package in its place she gets very excited and claps for herself. We all need to do things that give us a sense of accomplishment and Angelman individuals are no exception. The key is finding an activity that motivates them then making sure they are successful.

Debra, debradobrez@att.net, angel Megan, age 37, Del +

Having the same daily schedule is huge for our son. Especially naps and bedtime routine. If the schedule is off by a few hours he gets more anxiety and becomes restless.

AS Family Member

S

SAFETY

Special Needs Safety

Seatbelt/Strap Identifier

Car seat, seat belt and backpack strap identifiers that list name and diagnosis. In case of an accident or emergency this vital information would be available to first responders.

Project Lifesaver

Citizens enrolled in Project Lifesaver wear a small personal transmitter around the wrist or ankle that emits an individualized tracking signal. If an enrolled client goes missing, the caregiver notifies their local Project Lifesaver agency, and a trained emergency team responds to the wanderer's area. Recovery times for PLI clients average 30 minutes — 95% less time than standard operations.

AngelSense

GPS tracking and voice monitoring products with alerts, maps, listen-in capabilities and more.

Angel Care Monitor

Monitors with features like under-the-mattress movement sensor pad, infrared camera (day & night vision), 2-way talk-back, Wi-Fi internet communication and more.

Houdini Stop

Chest Strap that prevents your child from taking their arms out of their harnessing.

Angelman Family Contributions: Safety

Consider a durable wall mounted gate for the top of stairs where your child with Angelman syndrome plays and/or sleeps. This is especially important if your child has seizures. This continues to be a recommendation into teen/adult years.

Kelly

I couldn't keep Kade in his seatbelt. I found this little piece of plastic heaven called The Buckle Boss. I have also had a French door installed in my kitchen. He can see in; we can see out; but it has a lock and a dead bolt on it for his safety. We use hotel latches for our entry doors.

Hollie

At one of the ASF Walks we were told that it's a good idea to call the police station and let them know you have a child with special needs for their files. They now know that our daughter is a wander risk. Buy the Buckle Boss online to help keep your child in their car seat.

Jason, angel Chloe

Little hook and eye locks on doors to pantries, bathrooms and closets help keep little angels out of food, water, and toys. Just make sure that they are low enough for other siblings to get to those places.

Michelle

Using a baby monitor at night helps alert to seizures or getting sick from reflux.

Jo Lynn

To keep our angel out of all of the other bedrooms, we put the locks on the outside of all of our doors. This way he cannot get into anything dangerous.

Rene

Everything in our home is locked. We don't have knickknacks around the house. Every door is locked, every cupboard is locked, and knobs are removed from the stove. The refrigerator is locked, and especially the bathroom is locked. Child locks were not strong enough to keep my angel out, so we had to use durable metal locks.

Stephanie, angel Jeremiah "JJ"

Keypad locks sold at hardware stores keep angels safe and out of trouble. There are ones available for regular indoor doors and deadbolts. We have installed them on our front and back doors. There are also locks on the pantry door, the garage door, the siblings' doors, the furnace room and the office. It is a great investment. Use a permanent black marker to label the code directly on the lock for easy use by the family.

Jim and Deb, angel Justin

Due to decreased motor planning and spatial awareness, individuals with AS are likely to have significant safety awareness issues. Ensure that your child has a safe place to play and work by decreasing clutter (especially on the floor) and eliminate visual distractions in places that may be difficult to negotiate. Also consider safety devices, such as gates at the top of stairs and fences outside.

Kelly

Our guy is very mobile, and very curious, so we describe him as a large toddler. He is into everything all the time so we have to keep everything "baby-proofed" and we put away the breakables. He will not leave the electronics alone, and is forever digging into the cupboards and the refrigerator. So, we have locks on all the bedroom, refrigerator and garage doors. We also have a special enclosure on his bed to keep him from wandering at night, and we have alarms on all the exits so we know when he is trying to go outside without supervision.

Terri

I have an adult son who will not stop messing around with our big screen TV. So, I put a (office desk) plastic mat in front of the TV. I flipped it over with the "spikes" facing up. This works for any area that you do not want the child or adult to go near. This really works well.

Corbin, angel Tyler

We noticed that our granddaughter had a tendency to rock when she sat on a chair. We were afraid that she would fall off the chair. To prevent this from happening, I mounted a regular chair on a platform which was a piece of wood about 3/4 of an inch thick. I then mounted four iron pipe fittings into which I placed the chair legs. After that I poured roofing glue into the fittings. Once hardened, the legs are permanently glued to the fittings. With the chair mounted on the wooden platform this way, our granddaughter can't rock because the chair is mounted on the platform and the weight of my granddaughter keeps the whole thing stable.

Patrick

I purchased a pill container and every week I put all of his medications in there for each day. Then, I don't have to figure out or try to remember what he needs to take. It helps especially when you're in a hurry. *Aina, angel Ra'Shawn, age 21*

SCHOOL

See E-Education

SCIENTIFIC ADVISORY COMMITTEE (SAC): <https://www.angelman.org/about/scientific-advisory-committee/>

The ASF Scientific Advisory Committee (SAC) is responsible for overseeing research grants funded by the ASF. The ASF SAC is currently comprised of Angelman syndrome researchers and professionals from both academia and industry, clinicians who work with AS patients, and experts from related fields such as psychology, communication and education. SAC members donate their time and talent to review all research applications submitted to the ASF for funding, work with ASF staff to conduct an annual Scientific Symposium, and work with the ASF on research collaboration. The ASF and the ASF SAC play a strong role in networking researchers and funding research that pursues promising avenues of discovery. ASF SAC membership is evaluated and renewed on two-year terms, ensuring that a range of voices and opinions are expressed during the research funding process.

ASF 2020 SAC:

Stormy Chamberlain, PhD SAC Chair

Stormy Chamberlain, Ph.D., is a widely published, 10-year researcher in the field of Angelman syndrome and UBE3A, who has given more than 30 talks and lectures about a variety of related topics. Dr. Chamberlain is currently an assistant professor in the University of Connecticut's Genetics and Developmental Biology department, and is assistant director for UConn's Graduate Program in Genetics and Developmental Biology.

Arthur Beaudet, MD

Dr. Beaudet received his M.D. degree from Yale, did pediatric residency training at Johns Hopkins, and was research associate at the National Institutes of Health before joining Baylor College of Medicine where he has remained to the present. Beaudet has made diverse contributions in the field of mammalian genetics and publishing over 250 original research articles. He has studied Angelman syndrome for many years, and his lab and the Wagstaff lab independently identified the Angelman gene as UBE3A in 1997. Dr. Beaudet is currently the Henry and Emma Distinguished Service Professor and Chair in the Department of Molecular and Human Genetics at Baylor College of Medicine in Houston.

Charles Williams, MD

Charles A. Williams, M.D. is a Professor Emeritus of Pediatrics and Genetics in the Department of Pediatrics, University of Florida. He is a board-certified pediatrician and clinical geneticist and has a special interest in neurogenetic disorders. He is a past recipient of the Harry and Audrey Angelman Award for Meritorious Service. The main focus of his research career has been in the study of Angelman syndrome. Since 1982 he has published many papers on the genetic and medical aspects of Angelman syndrome.

Ben Philpot, PhD

Dr. Ben Philpot is an Assistant Professor in Cell and Molecular Physiology at the University of North Carolina, and a member of the Neuroscience Center, the Neurobiology Curriculum, and the Neurodevelopmental Disorders Research Center. Prior to this appointment, he was a postdoctoral fellow in the laboratory of Dr. Mark Bear at Brown University and a Research Scientist at the

Massachusetts Institute of Technology. Dr. Philpot earned his Ph.D. in psychobiology in the laboratory of Dr. Peter Brunjes at the University of Virginia in 1997. Dr. Philpot is known for his work examining the mechanisms by which experience regulates synaptic plasticity in the brain. His work has demonstrated that UBE3A is required for experience-dependent maturation of the neocortex, and he continues to elucidate the synaptic basis of Angelman syndrome.

Dan Harvey, PhD

Dan Harvey was appointed to the ASF Board of Directors in 2012 and previously served as chair the ASF Scientific Advisory Committee. Dan has more than 20 years of experience in drug discovery research both in academia and the pharmaceutical industry. He became involved with the ASF after his son, Matthew, was diagnosed with Angelman syndrome in 1996. From 1997 to 2001, Dan was a member on the ASF Board of Directors and served as vice president from 1997 to 1999. From 1999 to 2003, he chaired the ASF Scientific Advisory Committee. Dan earned a B.A. in Chemistry from the University of California, Santa Barbara and he holds a Ph.D. in synthetic organic chemistry from Yale University and was previously a member of the faculty of the University of California, San Diego. During his academic career, he was a Kent Graduate Fellow, an American Cancer Society Junior Faculty Research Fellow and an Alfred P. Sloan Research Fellow.

Michael Ehlers, MD, PhD

Michael Ehlers is Chief Executive Officer at Limelight Bio. Previously, he was Executive Vice President for Research & Development at Biogen. Dr. Ehlers grew up rural Nebraska and earned his bachelor's degree in chemistry from Caltech. He holds M.D. and Ph.D. degrees from the Johns Hopkins University School of Medicine.

Jane Summers, PhD

Dr. Summers is an Assistant Professor in the Department of Psychiatry and Behavioural Neurosciences at McMaster University and is Clinical Director of behavioral therapy services for children with developmental disabilities at McMaster Children's Hospital. She has been a member of the Scientific Advisory Committee since 2006. Dr. Summers has published papers on sleep and behavior problems in children with Angelman syndrome and training parents to implement ABA teaching procedures with their children. She has received grants from the ASF to study the effectiveness of ABA-based approaches for teaching functional skills to children with Angelman syndrome and to develop a battery to assess children's learning, memory and motor performance.

Steven Katz, MD

Steve Katz served as an officer or director of the ASF organization since 1997. He has served as President, Vice President, Secretary/Treasurer and Immediate Past-President as well as serving as a director. Steve has served on numerous Foundation committees, represents the ASF as one of two ASF representatives on the Coalition of Patient Advocacy Groups for the Rare Disease Clinical Research Network, co-chaired the ASF Biennial Conference in Philadelphia in 1998, has volunteered for countless ASF projects and coordinated the ASF walk-a-thon in Philadelphia for four years. Steve was awarded the 2007 Harry and Audrey Angelman Award.

Wen-Hann Tan, MD

Dr. Tan is a clinical geneticist at Boston Children's Hospital who has been conducting clinical research studies on Angelman syndrome since 2006 as a site PI of the natural history study and a few clinical trials. He has a broad interest in all genetic syndromes in children and adults, including the different "Angelman-like" disorders. He is actively involved in the development of novel outcome measures for

use in Angelman syndrome clinical studies. He is also actively involved in clinical trials in children and adults with inborn errors of metabolism.

Mark Nespeca, MD

Dr. Mark Nespeca is board certified in pediatrics, child neurology and in clinical neurophysiology. He earned his medical degree from Case Western Reserve University. His postgraduate training included a pediatrics residency at the University Of Colorado Affiliated Hospitals, a neurology fellowship at the University Of Utah Affiliated Hospitals and a fellowship in epilepsy/clinical neurophysiology at the Cleveland Clinic Foundation. Dr. Nespeca is the head of the AS Clinic at Rady Children's Hospital and Director of the EEG Laboratory at Children's Hospital San Diego. His clinical interests include epilepsy and he serves as vice chairman of the San Diego County Epilepsy Society Professional Advisory Committee. He is the principal investigator for two current epilepsy research studies. Recent titles of published work include "Subdural Electrodes in Infants and Young Children" in the Journal of Epilepsy and "Vocal Cord Paralysis as a Presentation of Intrauterine Infection with Varicella-Zoster Virus" in Pediatrics.

Fred Pritzker

Fred Pritzker is a practicing attorney and the father of a young man with AS. He is a former President of the Angelman Syndrome Foundation and a long-time member of the ASF SAC. Mr. Pritzker has also played a role in the evolution of ASF scientific funding strategies and policies including the targeting of specific research topics and the groundwork for an Angelman syndrome research institute.

Ron Thibert, DO, MsPH

Dr. Thibert received his Master's degree in Epidemiology from the University Of Massachusetts School Of Public Health and his medical degree from the Kansas City University of Medicine and Biosciences. He did his General Pediatric training in the Henry Ford Health System in Detroit, MI, and then completed a Child Neurology residency at the Floating Hospital for Children and Children's Hospital Boston in Boston, MA. He then completed a two-year fellowship in Pediatric Epilepsy and Clinical Neurophysiology at Massachusetts General Hospital. Dr. Thibert is now on staff in the Pediatric Epilepsy Program at the Massachusetts General Hospital for Children where he is the head of the Angelman syndrome Clinic, coordinates the Pediatric Epilepsy Surgery Program, and is a faculty member at Harvard Medical School. His current research projects include a retrospective study assessing the natural history and treatment options for epilepsy in Angelman syndrome, a prospective study assessing the efficacy of the Low Glycemic Index Treatment for seizures in Angelman syndrome, and another study assessing the significance of EEG findings in Angelman syndrome.

Katharine Grugan, PhD

Katharine Grugan received her doctorate in cell and molecular biology from Northwestern University in 2007. She has more than 17 years of biomedical research experience focused on oncology, immunology and the development of antibody therapeutics. Since 2011, she has been a scientist at Johnson and Johnson in their pharmaceutical division. She became passionate about the AS research field upon the diagnosis of her 10 month old daughter in 2011. Katharine resides outside of Philadelphia with her husband Kevin and three children.

Lora Meerdo

Lora Meerdo received a BS in biology from Alderson Broaddus University and an MS in animal reproductive science from Utah State University. After completing her MS she was part of the USU team that worked with the University of Idaho to clone a mule. Lora is currently a home school mom to her

three children, she serves as a volunteer leader of a local support group for families that have an individual with a disability and works at the Willow Park Zoo in Logan, UT. Lora lives in Hyde Park, UT with her husband Andy and their children Zachary (del +), Corban and Rosalie.

Elizabeth Berry-Kravis MD, PhD

Elizabeth Berry-Kravis MD, PhD is a Professor of Pediatrics, Neurological Sciences, and Biochemistry at Rush University Medical Center in Chicago. She has directed the Fragile X Clinic and Research Program since 1991, providing care to over 600 patients with fragile X syndrome (FXS). She has studied medical issues, epilepsy and psychopharmacology in FXS, and done translational research in FXS including outcome measures and biomarkers, natural history, newborn screening, and clinical trials of new targeted treatments in FXS. Recently, Berry-Kravis has expanded clinical and translational work to other neurodevelopmental disorders including autism spectrum disorders, and single gene ASD models, including Phelan McDermid syndrome, Rett syndrome and Angelman syndrome. She also is working on translational research in rare neurogenetic disorders including Niemann-Pick type C, Batten disease, pantothenate kinase-associated neurodegeneration, and creatine transporter deficiency. She has received the NFXF Jarrett Cole Clinical Award, FRAXA Champion Award, AAN Sidney Carter Award in Child Neurology and John Merck Fund Sparkplug Award.

Rebecca Burdine, PhD

Rebecca Burdine is a faculty member in the Department of Molecular Biology at Princeton University. Her lab focuses on understanding the mechanisms that control left-right patterning and asymmetric organ morphogenesis. She was named the 44th Mallinckrodt Scholar for the Edward Mallinckrodt Jr. Foundation, and received a Scientist Development Career Award from the American Heart Association in 2003. She was elected as fellow to the American Association for the Advancement of Science (AAAS) in 2018. She is on the Editorial board for Cell Reports and Zebrafish, and regularly serves on grant review panels for the NIH and NSF. Dr. Burdine is a parent to a child with Angelman syndrome. She first served on the ASF scientific advisory committee in 2007 by invitation from Dr. Joe Wagstaff. She has previously served as Chief Scientific Officer for the Pitt-Hopkins Research Foundation and for the Foundation for Angelman syndrome Therapeutics. She is currently serving on the steering committee for the Angelman syndrome Natural History study and on the Clinical Trial Steering Committee for the STARS and NEPTUNE studies conducted by Ovid Therapeutics.

Joseph P Horrigan, MD

Joseph P. Horrigan, MD, is a pediatric neuropsychiatrist. He has specialized in the treatment of children with complex neurodevelopmental disorders for more than 30 years. In addition, he has 24 years of experience in the various roles associated with the pharmaceutical and biotech industries, both as a clinical investigator and as a sponsor. Dr. Horrigan received his Sc.B. degree from Brown University and his medical degree from the University of Rochester. Dr. Horrigan has been a longstanding scientific advisor to FRAXA, he is a newly-elected Board member of Rettsyndrome.org, and he is also a Consulting Associate Professor at the Duke University Center for Autism and Brain Development in Durham, North Carolina.

SCIENTIFIC SYMPOSIA

See C-Conferences

The ASF Scientific Symposium is held every year. This two-day symposium is a chance for leading researchers, scientists and doctors to discuss the latest research activities in the world of AS. The first

day is focused on a specific topic with 5-8 presenters. This day is a unique opportunity for researchers to present their latest findings of their works and compare notes with colleagues from around the globe. The second day includes short presentations on all areas of AS.

SCOLIOSIS

[Click here to access the American Journal of Medical Genetics paper.](#)

Anna M. Larson Julianna E. Shinnick Elias A. Shaaya Elizabeth A. Thiele Ronald L. Thibert

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Scoliosis in Individuals with AS

- Affects 50% of individuals with an average age of diagnosis at 12 years old
- 24% of those diagnosed with scoliosis required surgery, an intervention disproportionately affecting men

<https://www.angelmanuk.org/>

[Click here for Information from the Scoliosis Association UK](#)

As children with Angelman syndrome age, progressive side-to-side curvature of the spine (scoliosis) may become apparent occurring in approximately 10% of children. The problem is more common in adulthood with about 40% of adult individuals with Angelman syndrome requiring braces or surgical correction. Scoliosis should be regularly monitored in all children and adults with the syndrome, particularly during periods of rapid growth (early childhood and adolescence).

Treatment Options:

The behavior of the curve may be monitored via repeated clinic visits and x-ray examinations at various times during development for worsening or progression of the scoliosis. Should the curve progress, or if it increases, treatment may be appropriate.

Bracing / Casting:

Bracing or casting programs may help delay or avoid the need for surgery. Bracing or casting congenital scoliosis (one present from birth) is rarely effective, but bracing or casting a small curve appearing during growth spurts curve may be helpful.

Bracing is prescribed depending on the degree of curvature determined by X-ray. The goal of bracing is to slow the progression of the curve, allowing the child to grow before a surgical procedure is done, as bracing alone rarely permanently corrects scoliosis.

Casting is usually used in cases of congenital scoliosis for children under the age of 3. It has been shown to either correct or to delay the progression of curvature in some cases, especially in young children and those with smaller curvatures.

Manipulation, physical therapy and/or exercise has not been shown to influence spinal curvature. Patients treated with scoliosis casting.

Surgery:

Surgery is generally recommended if brace or cast treatment fails to keep the scoliosis from progressing, or if the curve pattern shows brace or cast treatments are unlikely to succeed.

The surgeon will stop the progression of a curve without adversely affecting future growth. Various growth-friendly surgeries allow the spine and lungs to grow while controlling spine and lung deformity. For more information, visit Scoliosis Association UK <https://www.sauk.org.uk/>

SEIZURES AND SEIZURE MEDICATION

Information provided by the 2009 document written by Charles A. Williams, M.D; and Sarika U. Peters, Ph.D. [http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20AS%202009%203-19-10.pdf)

More than 90% of individuals with AS are reported to have seizures but this may be an overestimate because medical reports tend to dwell on the more severe cases. Less than 25% develop seizures before 12 months of age. Most have onset before 3 years, but occurrence in older children or in teenagers is not exceptional. The seizures can be of any type (i.e. major motor involving jerking of all extremities; absence type involving brief periods of lack of awareness), and may require multiple anticonvulsant medications. Seizures may be difficult to recognize or distinguish from the child's usual tremulousness, hyperkinetic limb movements or attention deficits. The typical EEG is often more abnormal than expected from the clinical appearance, and it may suggest seizures when in fact there are none [36, 61, 62].

There is no agreement as to the optimal seizure medication although valproic acid (Depakote), topiramate (Topamax), lamotrigine (Lamictal), levetiracetam (Keppra), and clonazepam (Klonopin) are more commonly used in the North America. Carbamazepine (Tegretol), ethosuximide (Zarontin), phenytoin (Dilantin), phenobarbital, and ACTH are less commonly used. Vigabatrin (Sabril), an inhibitor of GABA metabolism, should not be used. [63] Single medication use is preferred but seizure breakthrough is common. Some children with uncontrollable seizures have been placed on a ketogenic diet, and this may be helpful in some cases. Children with AS are at risk for medication over--treatment because their movement abnormalities or attention deficits can be mistaken for seizures and because EEG abnormalities can persist even when seizures are controlled.

Angelman Syndrome: Epilepsy and its Treatment for Providers

The Angelman Syndrome Foundation gratefully acknowledges the contributions to this publication: Ronald L. Thibert DO, MSPH(1); Kerry D. Conant(1); Eileen K. Braun(2); Patricia Bruno RN(1), Rana R. Said, MD(3); Mark P. Nespeca, MD(4); Elizabeth A. Thiele, MD, PhD(1)

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Angelman syndrome (AS) is a neurodevelopmental genetic disorder characterized by global developmental delays, severe speech impairment, disorders of balance or movement (usually ataxia), and frequent laughter, resulting from a defect in the maternally inherited copy of chromosome 15q11-13. A.S. can result from a deletion of this portion of the chromosome, inheritance of two paternal copies (uniparental disomy), a UBE3A mutation or a defect in the imprinting center. Some individuals meet clinical criteria for AS but do not have a clear genetic diagnosis. Epilepsy is present in over 80% of affected individuals, often presenting with multiple seizure types and is typically refractory to multiple medications.

Approximately 1,000 families of individuals with AS were contacted through the Angelman Syndrome Foundation (ASF) and asked to complete a questionnaire survey online. The survey contained detailed questions relating to the presence and presentation of epilepsy in AS, genetic subtypes of AS, and progression of epilepsy across the lifespan. Included were free text questions that asked respondents to describe their family member’s seizures in detail. The questionnaire additionally included detailed questions regarding the effects of various epilepsy treatments, both pharmacologic and non-pharmacologic, including free text questions asking family members to detail medication side effects. There were responses from family members of 461 individuals with AS (subjects included in the study). The subjects had an average age of 5.3 years (<1-35 years) at diagnosis, with 65% of subjects having a maternal deletion, 18% with an unknown subtype, 7% each with uniparental disomy (UPD) and UBE3A mutations, and 2% with an imprinting defect (ID).

Of the 461 subjects, 86% had experienced seizures with an average age of seizure onset of 2.9 years. Multiple seizure types were reported in 60% of subjects (average of 1.9 types), and the most frequently reported were atonic seizures (41%), generalized tonic-clonic seizures (40%), and atypical absence seizures (37%).

Approximately 32% were reported to have complex partial seizures and 6% simple focal motor seizures. Of those reported to have seizures with partial onsets, 8% had secondary generalization. Overall, 11% were reported to have only partial onset seizures, while 30% had both partial and generalized seizures. In addition, myoclonic seizures, tonic seizures, infantile spasms, and Lennox-Gastaut Syndrome were also reported (Table 1). Seizure types were determined by detailed free-text descriptions of the seizures provided by family members.

Table 1. Prevalence and frequency of various seizure types and syndromes in those with epilepsy due to Angelman syndrome.

Seizure Type/Syndrome	Prevalence	Frequency (seizures/week)
Atonic	41%	21.5
Generalized Tonic-Clonic	40%	9.2
Absence	37%	13.9
Complex Partial	32%	7.9
Myoclonic	12%	18.1
Tonic	9%	10.4
Secondarily Generalized	8%	8.6
Partial/Focal Motor	6%	11.4
Infantile Spasms	2%	12.3
Lennox-Gastaut Syndrome	1%	—

At the time of the survey, 34% were reported to be seizure free for a median period of 3.2 years with 23% experiencing seizure freedom for over 1 year. The average age of seizure freedom was 8.8 years. Of

the 396 subjects with seizures, 280 provided adequate data to determine rates of current epilepsy (those with seizure activity over the past year were considered to have “current” epilepsy). These rates are listed in Table 2. In addition, approximately 28% of those over 15 years of age reported an increase in seizure frequency after puberty.

Table 2. Percentages of those in various age groups with current epilepsy (as defined by seizures in the past year) for the 280/396 with epilepsy who provided adequate data as well as the 65 who never had seizures (N=345).

Ages	Percent with Current Seizures	Total in each Group (N)
<3	46%	30
3-5	60%	58
6-8	61%	51
9-11	71%	42
12-14	52%	46
15-17	53%	45
18 or older	59%	73

While it is unclear what percentage of subjects experienced non-convulsive status epilepticus (NCSE), 137 of the 396 with epilepsy (35%) were described as having some regressions in development. Of those instances of regression, 96 were attributed to seizure activity while 15 were attributed to medication and the remaining 26 to medical illness, environmental changes, or an uncertain etiology.

There were no statistically significant differences in seizure types or epilepsy rates based on gender, but there were clear differences in rates of epilepsy among genetic subtypes. Those with maternal deletions and unknown subtypes had the highest rates of epilepsy (89% and 90% respectively) while those with ID were the least affected (55%) (Table 3). There were no significant differences in seizure types amongst the different genetic subtypes, except for the more catastrophic epilepsies such as infantile spasms and Lennox-Gastaut Syndrome only occurring in those with deletions or unknown subtypes (Table 3).

Table 3. Prevalence of various genetic subtypes in Angelman syndrome with associated rates of epilepsy, multiple seizure types, and catastrophic epilepsy syndromes.

Genetic Mutation	Percent with AS	Percent with Epilepsy	Multiple Seizure Types	IS or LGS
Maternal Deletion	65%	89%	72%	4%
Unknown/Clinical	18%	90%	60%	3%

Uniparental Disomy	7%	75%	38%	0%
UBE3A Mutation	7%	74%	80%	0%
Imprinting Defect	2%	55%	80%	0

1. IS – infantile spasms 2. LGS – Lennox-Gastaut Syndrome

The most commonly prescribed medications amongst subjects with epilepsy were valproic acid (VPA – 62%), clonazepam (CZP – 34%), phenobarbital (PB – 30%), topiramate (TPM – 30%), carbamazepine (CBZ – 24%), lamotrigine (LTG – 24%), and levetiracetam (LEV – 20%). The complete list of medications is located in Table 4 along with average doses and lengths of treatment for the most commonly used medications. At the time of the study, subjects were on an average of 1.2 current medications, with 40% currently on monotherapy and 64% having tried multiple medications (average of 3.2 medications). Only 15% achieved good seizure control with their initial AED and an additional 8% with a second agent, with the remaining 77% refractory to medication.

Table 4. Percentage of individuals with AS and epilepsy who have tried various anti-epileptic drugs (AEDs), with average dose (mg/kg/day) and length of treatment for whom that information was available.

Medication	Percent Tried	Average Dose (mg/kg/day)	Average Course of Treatment
Valproic Acid	62%	16 (N=86)	51 months
Clonazepam	34%	0.4 (N=40)	36 months
Phenobarbital	30%	31. (N=9)	14 months
Topiramate	30%	4.4 (N=28)	30 months
Carbamazepine	24%	7.3 (N=3)	33 months
Lamotrigine	24%	8.1 (N=24)	13 months
Levetiracetam	20%	44.4 (N=23)	20 months
Phenytoin	20%	—	—
Zonisamide	10%	—	—
Ethosuxamide	8%	—	—
Gabapentin	7%	—	—
Felbamate	7%	—	—
Oxcarbazepine	5%	—	—

Tranxene	4%	—	—
Clobazam	4%	—	—
ACTH	2%	—	—
Nitrazepam	2%	—	—
Other	5%	—	—

1. “Other” includes pregabalin, mysoline, and vigabatrin.

Subjects’ family members were asked which medications worked best in controlling their epilepsy if they had tried multiple medications. VPA (25%) had the highest response rate followed by LEV (18%), LTG (17%), and TPM (14%). The lowest response rates were to CBZ (2%) and PB (2%). Similarly, LEV (37%) and VPA (28%) were associated with the highest rates of seizure freedom, followed by CZP (24%) and TPM (20%), with CBZ (4%) having the lowest rate. CBZ, by far, was associated with the highest rate of seizure exacerbation (59%), followed by PB (15%). See table 5 for a complete list.

Table 5. Efficacy of the 7 most commonly prescribed medications as evidenced by the percent that felt the medication worked best for them (of those who tried multiple medications), as well as those who felt the medication provided a period of seizure freedom or exacerbation.

Medication	Worked Best	Seizure Freedom	Seizure Exacerbation
Valproic acid	25%	28%	5%
Clonazepam	11%	24%	5%
Phenobarbital	2%	13%	15%
Topiramate	14%	20%	8%
Carbamazepine	2%	4%	59%
Lamotrigine	17%	11%	13%
Levetiracetam	18%	37%	12%

Rates of seizure freedom and seizure exacerbation for the most commonly prescribed AED in AS
Of the seven most commonly prescribed medications, approximately 50% or more of subjects who had tried CZP (64%), LEV (59%), VPA (54%), LTG (50%), and TPM (49%) were still on that medication at the time of the survey, whereas only 13% of those who had tried PB and 9% of those who had tried CBZ were still on those medications at the time of the survey, indicating these medications may have better tolerability and efficacy than PB and CBZ. Similarly, CBZ (45%) and PB (32%) were most frequently associated with intolerable side effects, followed by TPM (22%), VPA (17%), LTG (17%), LEV (14%) and CZP (5%). VPA did, however, have some potentially serious side effects with 3 patients reporting pancreatitis, 4 patients temporarily losing the ability to ambulate, 5 experiencing a drop in platelets, and one other experiencing a decreased white blood cell count There was also one child on LTG who developed Stevens-Johnson syndrome. See Table 6 for a complete list as well as the most common side effects for each medication.

Table 6. Tolerability of the 7 most commonly prescribed medications as evidenced by the percent still taking each medication and the percent that reported intolerable side effects, with the most common side effects listed for each medication.

Medication	Percent still taking	Worst side effects	Other side effects	Most common side effects
Valproic acid	54%	13%	4%	Tremor (8%); Fatigue (7%)
Clonazepam	64%	4%	<1%	Fatigue (8%); Hypotonia (6%)
Phenobarbital	13%	20%	12%	Lethargy (14%); Irritability (9%)
Topiramate	49%	15%	7%	Weight loss (8%); Cognitive slowing (7%)
Carbamazepine	9%	37%	8%	Increased seizures (20%)*; Fatigue (6%)
Lamotrigine	50%	12%	5%	Rash (6%); Fatigue (5%)
Levetiracetam	59%	10%	4%	Lethargy (5%); Irritability (4%)

Percentages of those still taking each of the most commonly prescribed AED in AS well as percentages of those who felt each AED had the most intolerable side effects.

Approximately 17% of subjects tried non-pharmacologic therapies for their epilepsies. The most common was dietary therapy with 40 subjects (11%) having tried this modality including 31 (8%) on the classic ketogenic diet, 7 (2%) on the low glycemic index treatment (LGIT), and 2 (~1%) on non-standardized diets. In addition to dietary therapies, 16 (4%) subjects had a vagus nerve stimulator (VNS) implanted. (Table 7).

Table 7. Efficacy and tolerability of non-pharmacologic treatments for epilepsy in AS as evidenced by the percent of those for whom the treatment worked best and the percent still using each treatment.

Treatments	Percent Tried	Worked Best	Still Using
Ketogenic Diet	8%	36%	19%

Low Glycemic Index Therapy	2%	0%	57%
Vagal Nerve Stimulator	4%	17%	50%

This is the largest study to date examining epilepsy and its treatments in AS. Epilepsy is very common in AS and, typically, quite refractory to medication.

Although epilepsy in AS is considered a generalized epilepsy, apparent partial onset seizures were fairly prevalent, though they did not respond well to medications, such as CBZ, which are used to treat focal seizures and typically exacerbate generalized epilepsies. Epilepsy was most severe in those with maternal deletions but, interestingly, the 18% of subjects who had an unknown/clinical diagnoses had epilepsy rates similar to those with deletions (90%), and the more catastrophic epilepsies such as infantile spasms and Lennox-Gastaut syndrome were only seen in those with deletions or unknown subtypes. Another significant finding of this study is that newer AED's, specifically LEV and LTG, and to a lesser extent TPM, appear to have similar efficacies in treating epilepsy in AS as compared to the older, more commonly prescribed medications (VPA, CZP) and have similar or possibly better side effect profiles with no need for routine blood monitoring (as with VPA), and less risk of potentially serious side effects. Non-pharmacologic therapies such as dietary therapy and VNS also show favorable efficacy and tolerability, but due to small sample size further studies are needed. Further characterization of epilepsy in AS, in addition to advances in genetic analyses, will hopefully lead to a better understanding of the pathogenesis of epilepsy in this population and, ultimately, better approaches to effectively treat epilepsy in AS.

Other websites about epilepsy:

<http://www2.massgeneral.org/childhoodepilepsy/index.htm>

This website provides education about epilepsy and its impact on children and families. This site includes video explanations of the different types of seizures, by Dr. Elizabeth Thiele.

<https://www.seizuretracker.com/>

This site is dedicated to providing people living with epilepsy and their doctors with free comprehensive tools to help understand relationships between seizure activity and anti-epileptic medications. Become active leaders in the treatment of your individual with AS, working hand-in-hand with doctors. Easy-to-use tools found at SeizureTracker.com allow you to create personalized reports of logged seizure activity and medication history that can be easily shared with their medical team.

Angelman Family Contribution: Seizures

I think there should be a topic listed for Shaking Angels. We have a Shaking Angel Group that has grown in 9 years to 315 members. It appears many of our loved Angels start these shaking episodes after puberty. Medications are hard to find to treat these episodes. Episodes do not show up as seizures on an EEG.

Mary Jo, mjkamp@cox.net, angel Katie, age 31, Del +

SEIZURE RESOURCES

Massachusetts General Hospital, Dr. Thibert, Angelman Syndrome Program

<https://www.massgeneral.org/children/angelman-syndrome/angelman-syndrome-program>

This website provides education about epilepsy and its impact on children and families. This site includes

videos of family stories as well as information on diagnosis and treatment.

SeizureTracker.com

SeizureTracker.com is dedicated to providing people living with epilepsy and their doctors with free comprehensive tools to help understand relationships between seizure activity and anti-epileptic medications. Become active leaders in the treatment of your individual with AS, working hand-in-hand with doctors. Easy-to-use tools found at SeizureTracker.com allow you to create personalized reports of logged seizure activity and medication history that can be easily shared with their medical team.

SAMi

Sleep Activity Monitor with monitors abnormal movements with a loud alarm, see and hear what is happening and makes recordings that you can share with your doctors.

Embrace

Smart watch that monitors behavior and sleep with alerts for unusual events.

Birdhouse Track Seizures app

Helps identify seizure triggers, keep a medication log and evaluate diets.

Low-Glycemic Index Treatment (LGIT) / Ketogenic Diet Resources

LGIT is a high-fat, limited-carbohydrate diet proven successful in reducing seizures in AS by up to 90%. Supervision by physician familiar with the treatment is needed.

Angelman syndrome Clinic at Massachusetts General Hospital

Where a group led by Dr. Ron Thibert, D.O., neurologist and pediatrician at Massachusetts General Hospital, completed a clinical research study on LGIT.

The Charlie Foundation

Provides resources and information about diet therapies including recipes, kitchen products and a worldwide list of locations where families can see a physician to be placed on the ketogenic diet.

SENSORY INTEGRATION

Angelman Family Contributions: Sensory Integration

Believe in sensory integration therapy. It works! Find an expert!

Terry, angel Byron, age 35

SHOES

Angelman Family Contributions: Shoes

After a lot of trial and error, we discovered this brand of shoes that fit nicely over our daughter's (3yo, del+) AFOs and now SMOs. The one Velcro strap is nice (so you can get them on quickly!). The *ikiki's* are also very lightweight, you are able to order them as singletons in different sizes, and there is an on/off switch for heel squeakers (for endless entertainment!). <https://ikiki.co/pages/ikiki-mobility>

AS Family Member

Our daughter is a “Houdini” when it comes to shoes. For years, the only thing that would work was lightweight high top wrestling shoes. When those became harder to find, we became desperate and tried Under Armor Kickit 2 Splatter mid tops. They are extremely lightweight, so she accepted them. However... she stills has “phases” when she pulls them off. It becomes a “fight or flight” situation and we believe she would break her ankle in order to succeed. Patience is the key!

Alice, angel Whitney, age 38, Del+ Class 1

SIBLINGS

Sibling Support Project

The Sibling Support Project is a national effort dedicated to the life-long concerns of brothers and sisters of people who have special health, developmental, or mental health concerns. The purpose of the Sibling Support Project is to increase the peer support and information opportunities for brothers and sisters of people with special needs and to increase parents’ and providers’ understanding of sibling issues.



Facebook Group for Adult Siblings:

<https://www.facebook.com/groups/902698506768244/>

When I Realized My Older Sister is Different

By: Christina Spaeth, Angelman syndrome sibling

<http://themighty.com/2014/11/when-i-realized-my-older-sister-is-different/> 11-20-2014

When I was a kid, I didn’t want adventure. I wanted normalcy. There were so many times I wished, for my sister’s sake, she could be a normal big sister who could give me advice, date boys and share clothes with me. That’s not what I have. I have Emily.

Emily has a random mutation of a single gene — something minuscule changed her entire brain, resulting in Angelman syndrome, a rare neuro-genetic disorder that causes severe developmental delays, absence of speech, gait/movement/balance problems and in most cases, seizures. We’re lucky, however, because unlike the majority of cases, Emily has a mutation of the UBE3A gene, instead of the deletion.

She’s therefore more mildly affected than those with the deletion, who sometimes cannot walk. Emily learned to walk at the age of 3 and has never had seizures. Because the technology was not yet developed enough to detect her genetic mutation when she was young, she was not diagnosed until high school after extensive genetic testing at the University of Chicago. So before that, I had to use phrases like “severely cognitively disabled” in an attempt to explain my sister.

I remember the day I realized my sister was different. I was around 3. I’d gotten a splinter and ran inside to my mom. I sat down on the kitchen floor and waited for my mom to get tweezers and a Band-Aid. Emily, 6, sat down next to me and fussed for a Band-Aid. “I know, honey. You want one too,” my mom said, wrapping one around Emily’s finger, even though there was no need. As I sat there watching this, some connection sparked in my brain: “I’m growing up, and Emily never will.” That is one of my first memories.

Emily turned 23 this year. We have another brother who is five years older than her. It was a confusing limbo for me as a child, since I was physically younger than Emily but mentally older. Emily will always be about 3 years old cognitively. When I was a kid that was kind of fun since I always had a playmate. We would run around in the backyard (albeit, Emily kind of shuffled) and swing and play in the sandbox. I learned not to get upset when she knocked down my sandcastle and to stay away if she got angry. As my parents taught me, the rest of us have the ability to use words when we're upset, but Emily doesn't so she expresses it any way she can, generally by having a tantrum. The best thing to do was get out of her way when she was mad and let my parents handle it.

I didn't know how to talk about my sister when I was young. One neighbor girl, when we were 5, asked, "Does your sister wear diapers?" I hated the snotty look on her face so I lied and said no. A few years later, at Chuck-E-Cheese for my eighth birthday, Emily and I were crawling around the tubes and two younger girls watched as she passed by. "She's fat," one said to her friend. I bit my tongue and shoved Emily forward. Even though she didn't understand the words, I certainly did. In middle school, when a fellow classmate was making fun of individuals with disabilities, I kicked him in the shin.

All of this information, these anecdotes, these memories, have been buzzing around my head for months. I've been trying to organize my thoughts about Emily for a long time. How does she see the world? How does she see me? I'm her translator, her referential point in new situations, her protector, her teddy bear. I know her language of noises, pointing and signs as if I were bilingual. It's not easy all the time. She has impressive tantrums, sometimes in public.

She's like a clingy puppy when I'm home from college. And yes, she can be kind of gross, like a toddler would be. I'd rather not say the amount of times I've been sneezed on.

But you know what? Without her, I wouldn't be who I am. I wouldn't be as compassionate and patient towards people of all abilities. I wouldn't have written about her in my college essays and in magazines. I wouldn't be studying to be a speech-language pathologist.

Emily is always giving hugs and kisses. Yes, she has big temper tantrums, but she generally has an exceptionally happy demeanor. She has the biggest belly laugh you ever heard. She loves things with wheels, musicals, bowling, baseball, swimming, ice cream, family and friends. Her vision and hearing are sharp and her memory even more so. She never forgets a face or a route to get somewhere. She adores riding in the car and could do it for hours on end. She loves to cuddle. She likes to tease and play jokes on people. She writes "Em" on everything she owns. She squirrels away things in her many backpacks, stuffing them beyond belief. She enjoys shredding paper. She flaps her arms when she's happy. She cries at sad music and laughs at slapstick comedy. She's the most loving, forgiving person I know. That is what I have. I have Emily.

Angelman Family Contributions: Siblings

Siblings of children with Angelman syndrome face unique challenges. Many siblings often offer their help. However, it is important for parents to encourage siblings to spend time with their peers and participate in extracurricular activities. Sibshops and peer support groups can be a fun and social way for siblings to deal with their feelings about AS brothers or sisters. Many siblings have reported feeling more independent and sensitive because of their angel brother or sister.

AS Family Member

Concerning having more children, the most common worry parents feel is that they won't have enough time to spend with typical children because of the care required by their AS child. They also express concern about the future and leaving other siblings with too much responsibility after they are gone. However, one mother reported that having a second child was the best thing that ever happened because she appreciated every tiny milestone and was in awe of a typical developing child. Although each child and situation is different, many sibling or relative's thoughts and feelings are similar. The absence of speech seems to be one of the biggest issues for children to understand and accept. The most important thing to keep in mind is that children are bound to experience negative thoughts and feelings towards their Angelman siblings at different times throughout their lives. The feelings are normal and should be respected and discussed. Dealing with feelings of anger, resentment and disappointment early on can help lay the groundwork for a strong and positive relationship between your children later in life. Children need to know it is okay to experience these feelings and you will be amazed at the response you get when you tell them that you have experienced these very same feelings, as well.

AS Family Member

My daughter Elena, age 23, is living at home with her father, mother and two of her three siblings. She is the second of four siblings. When her brother Daniel was born, she was seven years old. When we got Daniel home from the hospital, the baby was crying inconsolably. Elena took a piece of plastic that apparently she had hidden under the couch and put it over his crib in an effort to calm the crying baby. Elena was giving him his greatest treasure...her piece of plastic!! This was her first expression of love for her little brother.

Maria, angel Elena

We suggest including your angel in sibling sports activities. Justin is the flag bearer and equipment manager for the football team. He led the team onto the field for games and helped carry the equipment for practices. It's been such an incredible experience. The boys on the team are from all over the city and are always very excited to see Justin. He is treated like a celebrity and loves all the attention. We now have a lot more people knowing Justin. He is accepted and there is a greater understanding of Angelman syndrome throughout the community. It's a win-win situation for both our son and his peers.

Jim and Deb, angel Justin

Our older daughter said it best when she exclaimed, "There is no normal! Everyone is different and needs to be cherished." She loves her little sister with all her might, but her little sis can still drive her crazy! I tell her that THAT is normal! Siblings can and will drive you crazy. That's their mission in life, but they can also be your best friend!

Leah, angel Emily, Del +

Both of our angel's siblings say he has teaching them to be patient and to value what really matters...not the material things. They also have said that having an angel as a sibling has helped the family to be more united.

Brisia, angel Mario

Our angel attended her older sister's sports activities and essentially became a "mascot". It taught our daughter's friends invaluable lessons about inclusion and sensitivity toward others with special needs. It also greatly enhanced the life of our angel and she loved being part of all of the action!

Alice and Mark, angel Whitney, age 38, Del+ Class I

We used to feel like we weren't able to do enough for Jennie's older brother, Jim. However, he turned out just fine! My husband and I would take turns taking our typical child special places and made sure he had friends over and went to their houses. He was in lots of local recreational activities, too. I would also try to give him a special time every day. At a recent family birthday party, instead of running around with the other kids, Amanda, (my AS son, Devin's cousin) brought a big pile of books downstairs and spent the next half an hour on the couch "reading" to Devin. *AS Family member*

I had four teenagers in junior and senior high when Susan (AS) was in the seven to ten year-old age range. I think it all balanced out very well. Their problems at times were so challenging that we couldn't get overly involved with Susan and her needs. At the same time, caring for her kept us from becoming over anxious about all those typical teens! I am pleased to report that our typical children seem to be well-adjusted young adults. They now joke about all the things they got away with because of Susan's needs!

AS Family Member

Samantha (AS) has an older brother who is seven and a younger one who is almost three. We really didn't think about having another child, it just happened. It has been the best thing for Sam. They all do a lot together and she really watches her younger sibling. The younger one has surpassed her developmentally and so now he is a good role model. They fight like cats and dogs sometimes, just like any siblings would. He has begun talking for Sam and is very protective of her. He is always saying, "Mom, Sam wants...." They love to wrestle and watch Sesame Street together and were even holding hands in the back seat of the car the other day. Her older brother also loves to wrestle with her and he is excited that she will be in his school next year. They do fight more because he wants more space and she loves to invade it!!! But, he is very protective of her. When he was little he responded to an explanation of why Sam couldn't talk by saying that he knew what the problem was – Sam was talking, but nobody understood her because she was speaking French! For us, it can be hectic, but having other children allows us not to focus too much on Samantha. She does need a lot of care, but having other children allows us to focus on the more ordinary things in life. So, it can be a difficult decision but I think it is very beneficial to have other children.

AS Family Member

When Ashleigh (our older daughter and AS sibling) was six we enrolled her in the Big Brother/Big Sister program. It was the first time the local group had a child who wasn't from a single parent family. I explained that I wanted a big sister for Ashleigh because I felt she could use one-on-one attention, and because I wanted her to have a "sibling" she could talk to and someone she could share a special bond. Her first Big Sister was a television news editor. Ashleigh was in her wedding! Her second Big Sister, who was a marriage and family counselor, spent many years with her and attended her high school graduation. It was so wonderful for Ashleigh. They went to the beach, the movies, rollerblading, shopping, etc.

Alice and Mark, angel Whitney, age 38, Del+ Class I

SLEEP CHALLENGES

Angelman Family Contributions: Sleep Challenges

Melatonin has been a life saver for our family. Esmae takes 3mg dissolvable a half hour before bedtime and it generally allows her to sleep approximately 10 hours each night.

Julie, julialouis@howardhanna.com, angel Esmae, age 5, UPD

When our daughter was about four months old, she kept aspirating liquids. We were able to get her scheduled for a supraglottoplasty - this is to help with her windpipe. The surgery was a huge success and the biggest surprise is now she can SLEEP THROUGH THE NIGHT!! Prior to this, we had HUGE sleep challenges, and then we got the surgery and there is a NIGHT AND DAY difference. She was a totally different kid. Cannot say enough about supraglottoplasty.

Alex, angel Rose, age 2, Del+

I do not know whether or not this behavior is common. But, my brother sleeps to the sound of other family members sleeping. So if he is restless, I pretend I am already asleep and I make a gentle snoring sound. If he was awake simply because he was restless, and not because of pains, this tip will make him sleep.

AS Family Member

Routine is essential for our Angelman child. It assists with normalizing his wake/sleep cycle and keeps his behaviors to a minimum if he knows what to expect as the day progresses.

Terri

A low dose of Clonidine better known as blood pressure medication. Clonidine was confirmed by my Angels Geneticist to have worked with other Angels. It helps to keep Angels asleep through the night.

AS Family Member

Establishing a routine for sleeping helped. We would all get into our own beds at the same time. Initially my son would get out of his bed and crawl around, but gradually he started staying in bed and sleeping through the night. In the initial phases twice under his pediatrician's supervision we gave him low dose melatonin tablets for a 3 month duration and weaned him off. It supported the initiation of the sleep through the night behavior.

AS Family Member

SOCIAL SECURITY AND SSI

Benefits <http://www.ssa.gov/pubs/EN-05-10026.pdf>

Social Security Disability Benefits & SSI Disability Claims

<http://www.disability-claims.net/>

Questions: Contact Dr. Eric Wright, ASF Family Resource team member, via an online form at:

<https://www.angelman.org/resources-education/asf-family-resource-team/>

SPECIAL NEEDS TRUST

Here is a website that offers further information:

<http://specialchildren.about.com/od/longtermplanning/qt/fftrust.htm>

A special needs trust is a trust designed for beneficiaries who are disabled, either physically or mentally. It is written so the beneficiary can enjoy the use of property that is held in the trust for his or her benefit, while at the same time allowing the beneficiary to receive essential needs-based government

benefits. There are administrative advantages of using a trust to hold and manage property intended for the benefit of the beneficiary if the beneficiary lacks the legal capacity to handle his or her own financial affairs.

In the United States, such trusts provide advantages in helping beneficiaries qualify for health care coverage under state Medicaid programs, and also for monthly cash payments under the Supplemental Security Income (SSI) program operated by the Social Security Administration.

Angelman Family Contributions: Special Needs Trust

Through a lawyer, we set up an irrevocable trust for our son. This is so we can provide funds for things he needs that Medicare and Medicaid will not pay for after our death.

Susan

STRAWS

See also D- Drinking

Practice drinking from a straw using box drinks with a straw that the liquid can be gently squeezed up the straw working on them trying to suck the liquid up the straw themselves. Once they can drink from a straw practice using oral muscles by drinking thicken liquids from a straw (you can make your own smoothies using applesauce with juice, or yogurt with milk, or pudding with milk)

Debbie, Occupational Therapist, Rady Children's Hospital, San Diego, CA

STROLLERS

Adaptive Mall:

https://www.adaptivemall.com/allstrolpus.html?msckid=e0a5e057677b148d5e42bd9b31180e6d&utm_source=bing&utm_medium=cpc&utm_campaign=ECI%20-%20Strollers&utm_term=Special%20Needs%20Jogging%20Stroller&utm_content=General

SUPPORT RESOURCES

Visit ASF website for links: <https://www.angelman.org/resources-education/support-resources/>

Ability Path

Support for Parents of Children with Special Needs.

Article: States of Grief for Parents of Children with Special Needs

Miracle Flights

Provides flight support for domestic or international travel to U.S. facilities for medical treatment, second opinions and follow-up. Domestic service dog training and retrieval and organ/blood travel assistance.

Project Lifesaver

Citizens enrolled in Project Lifesaver wear a small personal transmitter around the wrist or ankle that emits an individualized tracking signal. When notified that an enrolled client goes missing, a trained

emergency team responds to the wanderer's area. Recovery times for PLI clients average 30 minutes — 95% less time than standard operations.

National Disability Navigator

Cross-disability information and support for ensuring people with disabilities receive accurate information when selecting and enrolling in insurance through the Affordable Care Act Marketplaces.

What I Would Tell You

An inspirational firsthand account of raising an individual with a disability, by Julie Keon, who spent most of her professional life working as a Certified Doula. She is the mother to Meredith who was born full term but had an abrupt lack of oxygen at birth resulting in severe cerebral palsy.

Family TIES

Offers information and resources, parent-to-parent support, and workshops for families and professionals supporting children with special needs. (For Massachusetts residents)

The Air Care Alliance

Nationwide league of humanitarian flying organizations whose volunteer pilots are dedicated to community service.

SURGICAL PROCEDURES AND PREP

Information provided in 2009 document written by Charles A. Williams, M.D; Sarika U. Peters, Ph.D.
[http://www.angelman.org/_angelman/assets/File/facts about as 2009 3-19-10.pdf](http://www.angelman.org/_angelman/assets/File/facts%20about%20as%2009%203-19-10.pdf)

There are several literature reports of individuals with AS undergoing general anesthesia without any difficulties. Also, the experience reported by many parents on the web and from parent meetings is generally favorable regarding successful general anesthesia and other aspects of surgical intervention. Some scientific reports mention concern for those who have the deletion mechanism (present in 70% of those with AS) because these individuals also have a deletion of GABA receptor genes which are known to be targets of certain anesthetic agents such as benzodiazepines and halogenated ethers. However, the weight of experience thus far indicates that individuals with AS tolerate anesthetic agents well. Convalescence from surgical procedures also appears to occur relatively normally. For example, rather significant surgical interventions of scoliosis repair with rod replacement and bone grafting is not uncommon to be well tolerated in those with AS.

There have been recent reports of bradycardia in individuals with AS and the presumption has been that these rhythm problems were due to increased activity of the vagus nerve [71--73]. These reports are somewhat difficult to interpret because of the complexities associated with hospitalization such as multiple medication use and the variables of the surgical procedure. At this point, it seems unclear if individuals with AS have an increased risk for cardiac rhythm disturbances. Anesthesiologist should certainly be aware of these case reports however and of the possibility that agents that increase vagal tone may not be well tolerated in individuals with AS.

Angelman Family Contributions: Surgical Procedure

Our daughter has had several surgeries. First, our hospital (UCSD) has always let her wear her pajamas or other familiar and comfortable clothes. She has never worn a gown and she would likely never

tolerate it. We always have requested Zofran to prevent nausea and vomiting post-surgery. The last thing we want is for her to suffer from vomiting and be miserable after the procedure. Next, the hospital has always allowed one parent to put on scrubs and accompany her to the OR. The best method for us is to wheel her into the room in a chair and then lift her to the table. My husband usually is the one to go since he is stronger (physically and emotionally). He is there to help hold the mask until she falls asleep. He is face to face with her... and he always hopes he doesn't inhale and collapse! :)

Alice, angel Whitney, age 38, Del+ Class 1

SWIMMING

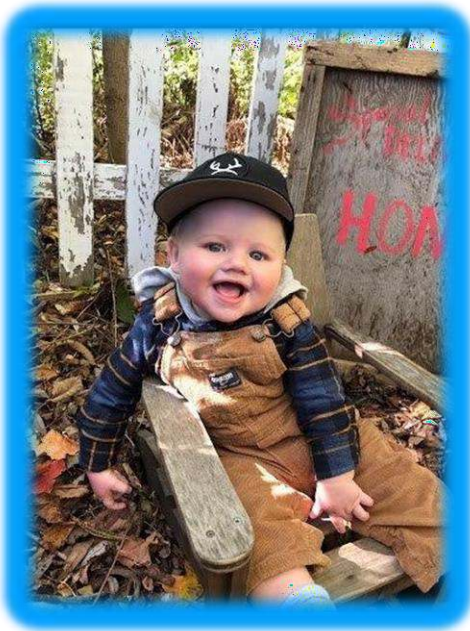
<http://www.sosecureproducts.com/>

Made by Discovery Trekking Outfitters. An effective and discreet unisex swimming undergarment for adults and teens.

Angelman Family Contributions: Swimming

Our daughter loves aquatic barbells! She loves holding them while she walks around the pool independently- with our supervision!

Alice, angel Whitney, age 38, Del+ Class 1



T

TESTING TO DETERMINE ANGELMAN SYNDROME

<https://www.ncbi.nlm.nih.gov/books/NBK1144/> Aditi I Dagli, MD, Jennifer Mueller, MS, CGC, and Charles A Williams, MD.

Method	Genetic Mechanism Detected 1					Total Proportion of AS Detected by Method 2
	15q11.2 -q13del	UPD	ID	UBE3 A seq	UBE3A del/dup	
DNA methylation analysis 3, 4	X	X	X 5			~80%
MS-MLPA 6	X	X	X			~80%
FISH 7	X					~68%
CMA 8	X	X 9				~68%
UPD study 10		X				~7%
AS IC deletion analysis 11, 12			X			~3%
UBE3A sequence analysis				X 13		~11% 14
UBE3A gene-targeted deletion/duplication analysis 11, 15					X	Rare

Possible explanations for the failure to detect AS-causing genetic abnormalities in the 11% or more of individuals with clinically diagnosed AS:

- Incorrect clinical diagnosis
- Undetected pathogenic variants in the regulatory region(s) of *UBE3A*
- Other unidentified mechanisms or *gene*(s) involved in *UBE3A* function

THEATHER

AMC Sensory Friendly Films <https://www.amctheatres.com/programs/sensory-friendly-films>

AMC Theaters offers sensory-friendly film showings to families affected by autism and related disorders, such as AS. See the schedule of upcoming films and participating AMC theaters.

THERAPIES

Music Therapy

Coast Music Therapy, Inc.

Coast Music Therapy is a San Diego-based agency committed to serving children and adolescents who require a creative approach to learning by utilizing music to facilitate individual growth and achievement.

Tuned Into Learning

Tuned in to Learning is a comprehensive music-assisted learning curriculum for special education.

American Music Therapy Association

Find a qualified music therapist in your area.

Physical Therapy

Therapies 4 Kids

Therapies 4 Kids is an intensive therapy program for children with neurological disorders who have physical disabilities and are in need of Pediatric Intensive Therapy.

Siblings

Sibling Support Project

The Sibling Support Project is a national effort dedicated to the life-long concerns of brothers and sisters of people who have special health, developmental, or mental health concerns. The purpose of the Sibling Support Project is to increase the peer support and information opportunities for brothers and sisters of people with special needs and to increase parents' and providers' understanding of sibling issues.

Angelman Family Contributions: Therapies

Start PROMPT therapy for apraxia as soon as you get a diagnosis!

<https://promptinstitute.com/general/custom.asp?page=PROMPTtraining> and

<https://www.kidstherapyassociates.com/hanen-prompt>

Carolina, tinyangel5@yahoo.com, angel Liora, age 10 UBE3A mutation

Therapeutic horseback riding has made an enormous difference in Rebekka. While she has ridden horses off and on during the past 20 years, about 6 years ago we connected with a local therapeutic riding program that has changed her physical abilities (she shifts items from one hand to another, crosses midline all the time now, can throw and catch a ball) to the point where we discontinued physical therapy! She has learned (some) patience and loves caring for the horse, Geronimo. While she still has no balance to walk, she sits straight on the horse and has a very strong core. I wish we had started this years ago!

Lynne, lcfort@coomcast.net, angel Rebekka, age 32, Mutation

I want to share something that our physical therapist told us that might be helpful to other Angelman families. During a session one day, I remarked that my son Jack was babbling more during PT than he does during his speech therapy sessions and she told us that movement encourages speech! Who knew! So if it possible to incorporate some sort of movement during speech therapy (rather than just sitting in a chair) that might encourage more babbling. I definitely find this to be true for our son Jack!
Sayoko, sayoko.murase@gmail.com, angel Jack, age 2, Del+

TOILETING

Applying Structured Teaching Principles to Toilet Training

How to create structure and routine to toilet training for a child with Autism – an article by Susan Boswell and Debbie Gray through the University of North Carolina TEACCH® Autism Program.

Angelman Family Contributions: Toileting

Big tip for this is TIMING. We take our daughter in the morning, after school, and at night. This regularity has helped her stay dry more than anything else.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

Toilet training may take a long time but if you are patient it is well worth it. We started when my son (Del +) was two years old. He had problems sitting, so we used a potty chair that sat on the floor with a back and arms. We determined a bowel movement schedule first. It was right after breakfast, so at that time we would sit him on the potty. We gave him books to look at and he would stay until a BM was produced. He would let us know when he had to go at other times (usually after a meal). Toilet training for urine took a bit longer. We kept a two-week schedule in which every potty of the day and all liquid intake was documented. The day care and school also documented while he was there. This was a lot more difficult, but we saw slow progress. He was “timed trained” but he still indicated at other times when he had to go. He had a sound for potty and when he made it we immediately took him. He was five years old before he was dry all night. It was well worth the effort! He still needs assistance with wiping, yet he always indicates and lets us know verbally when he has to go to the toilet. We still put a TV table in front of the toilet so he can read a magazine or book while on the toilet.

Coral, angel Trent

Gavin had a lot of anxiety over going into the bathroom. He would stand in the kitchen and pee in his diaper on demand before we went somewhere. He was ready to wear underwear, but he wouldn't go into the bathroom to pee. With him being a boy, we started having him pee into plastic cups in the kitchen. We took away the element of the bathroom that was giving him anxiety. He was very successful at peeing in the cup on demand. Soon after we started this he had to go poop. We then used two cups to catch the pee and poop. After about a week of peeing and pooping into cups in the kitchen, we moved into the bathroom and continued using two cups. After he was comfortable just being in the bathroom we had him stand in front of the toilet and pee into the toilet and still held a cup for the poop. It took a lot longer to get him to sit on the toilet to poop, but he eventually transitioned to it. I also kept cups in the car for a long time for when he had to go and couldn't hold it.

AS Family Member

Three (2 ASD, 1 AS) of my four boys have sensory issues, especially to loud noises. Getting them to use the toilet in public places was daunting to say the least until I found a trick that has works for us. I always carry a pack of sticky notes in my purse for automatic flushing toilets. We put a sticky note over the sensor and when my boys are done toileting they pull it off and throw it away. This way they are able to anticipate the flush and do not get scared by a loud flush mid-way through toileting.

AS Family Member

We attached a “bidet seat” to Kathryn’s commode. It had a seat warmer which helps her to relax and know to use the bathroom. It also makes it much easier to clean her.

Bill and Paula, angel Kathryn

We found it less stressful by practicing routine timed toilet training first. We bring our son to the restroom when he awakes in the morning, after meals, and before bed. For BM’s we just watch for facial expression and body movement.

Maria

As much as we would like to hurry the bathroom visits, we have concluded that it is best to accept the fact that it is going to take some time. We find ourselves quietly “meditating”- for lack of a better word- when our daughter is using the restroom. We look down, don’t make eye contact and WAIT! We have used “bartering” at times... “First, you go to the bathroom and then we’ll get up and eat...” But, truly the most effective technique for us is SILENCE AND PATIENCE!

AS Family Member

Our daughter’s school started “habit training” or timed training when our angel turned three. We thought they were crazy... they weren’t! They instructed us to send her in Pull-ups and we never went back. (We did continue diapers at night for a few years.) I believe the school put her on the toilet every two hours and we did as well. It proved that establishing a routine is a crucial learning tool for our children. Our daughter was later than some angels to reach other developmental milestones, but this was one area for which she excelled, and we owe it to her school and our diligence and bravery!

AS Family Member

We have come to accept that our angel will likely not use the toilet in a new or “strange” place. This often happens when traveling. We do worry about her holding urine for longer periods of time. Pushing liquids when traveling is important. Taking her out of her routine does have consequences, and this seems to be one. Eventually, she does adjust to the new surroundings.

AS Family Member

We bought a strap with bells on it that is used for dogs to alert their owners when they need to go outside. We hung it in the bathroom so our angel can ring the bells quickly and easily when she is ready to use the toilet. We can hear them from across the house and we come running.

AS Family Member

The system that helped us get our angel to learn to stay dry at night was the Rodgers bedwetting alarm training underwear. It had a battery pack sewn into the pants and detachable portion that could alert us at night to get our angel to the toilet when sensors detected wetness and then he would understand what sensation meant it was time to go potty. We would put disposable training pants on over the underwear so we had less mess until he started to get up to go to the toilet himself. These can be used

in the day, of course, as well.

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

TOOTH BRUSHING

Angelman Family Contributions: Tooth Brushing

We wrap our son in a large towel so he's cocooned inside. It means he can't hurt us or himself when he's struggling to stop it. *AS Family Member*

Brantley 2 (deletion +) loves brushing his teeth! Since he was smaller I would use a vibrating toothbrush. He's always so amused with it.

Sarah, j.sorreis09@blueriver.net, angel Brantley, age 2, Del+

Use of a Collis curve toothbrush with which you can brush all three surfaces of the tooth in one motion before biting starts. <https://www.kleenteeth.com/brands/Collis-curve.html?msclkid=de51090760ae1b9c76ea54c6aa4c22f6>

Dr. Tony, angel Sarah Nicole

Brushing your angel's teeth while in the shower or bath makes life so much easier!

Kathleen

We learned early on that the only way we could brush our angel's teeth is to wrap her in a blanket. We take our time, and she doesn't mind it, but those hands get in the way if we don't "burrito" her.

Sandy, angel Elizabeth, Del+

Tooth brushing is a sensory experience that can be overwhelming. Some things that help are: 1) letting my son play with the toothbrush for a minute before using it; 2) giving him a sensory toy to hold while I brush.

AS Family Member

TOYS

See P- Photo Books and Blankets

Special Needs Toys

Triad Theraplay

TRIAID is the manufacturer and supplier of special needs equipment for children, specializing in special needs tricycles and handcycles.

TFH Special Needs Toys

TFH Special Needs Toys present a line of carefully selected Special Needs Toys designed to help you or those in your care enjoy life, and achieve more.

FlagHouse

Flaghouse is a global supplier of physical education equipment and products, equipment and programs



to both physical education and recreation professionals, as well as professionals who deal with children and adults with physical and developmental disabilities.

Online Stores for Sensory and Adaptive Toys

Following is a list from the ASF Education Webinar with Lizzie Sordia on Products and Services

<u>Fat Brain Toys</u>	<u>Beyond Play</u>
<u>Warner’s Corner Toys</u>	<u>Special Needs Toys</u>
<u>Different Roads to Learning</u>	<u>Adaptive Mall</u>
<u>eSpecial Needs</u>	<u>Sensory Goods</u>
<u>Fun and Function</u>	<u>Child Therapy Toys</u>
<u>Playability Toys</u>	<u>Oompa.com</u>
<u>Chewy Tubes</u>	<u>Talk Tools – sensory tools</u>
<u>SentioChews</u>	

Sensory Toys on Google Express

Google Express is a delivery service powered by Google where you can shop from stores like Walmart, Target, Costco, Walgreens, PetSmart, and more.

The Ultimate Amazon Special Needs Shop by Angelman Today

A collection of Special Needs toys compiled by Angelman Today. When you make a purchase from the collection, your purchase supports *Angelman Today*.

2019 Holiday Gift Guide Amazon

https://gallery.mailchimp.com/3978984f5d484d7547e71477f/files/651d31d1-939c-4747-b720-ea90dc344700/2019_Holiday_Gift_Guide_s_.pdf

Angelman Family Contribution: Toys

Our son always enjoyed soft or sturdy talking toys and stretchy items, bath toys and also blocks that can be put in and out of containers, like a mailbox shape sorter, or simply Legos and large plastic containers!
Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

TRAVEL

Travel Resources

<https://www.angelman.org/resources-education/?cat=85&yr=undefined&kw=Research%20keyword>

Center for Courageous Kids

The Center for Courageous Kids, family retreat weekends are designed to provide respite, recreation, and support programs for families who have a child with a chronic or life threatening illness. Family weekends provide similar experiences as summer camp, as well as the opportunity for parents to gather together and share similar life-experiences.

Barren Heights Christian Retreat Center

Provides families that have children with a physical or developmental disabilities a free weekend retreat.

Discovery Cove

Discovery Cove offers accommodations for guests with disabilities and is located in Orlando, FL. Guests can experience a 30-minute Dolphin Interaction, snorkel with tropical fish and rays, wade beside marmosets and otters, hand-feed parrots, toucans, and other exotic birds in, and relax in the large resort pool and swim along a tropical river.

Give Kids the World

A 70-acre nonprofit “storybook” resort, located near Central Florida’s most beloved attractions, where children with left-threatening illnesses and their families are treated to week long, cost-free fantasy vacations.

Grandchildren with Special Needs: Where to Vacation

Discover destinations that are best for kids who need extra consideration.

Morgan’s Wonderland

Created by philanthropist Gordon Hartman inspired by his daughter Morgan, a child with special needs. A special place for special friends – an environment for inclusion and understanding – an oasis for those needing a safe place to relax and enjoy the outdoors.

Products, Services and Advice for Travel

Seatbelt/Strap Identifier

Car seat, seat belt and backpack strap identifiers that list name and diagnosis. In case of an accident or emergency this vital information would be available to first responders.

Miracle Flights

Provides flight support for domestic or international travel to U.S. facilities for medical treatment, second opinions and follow-up. Domestic service dog training and retrieval and organ/blood travel assistance.

RoadID

Road ID is a durable, rugged, athletic, fashionable line of identification gear. Currently, they offer five different forms of ID: the FIXX ID, the Shoe ID, the Ankle ID the Wrist ID and the Shoe Pouch ID. We provide various forms of ID to allow a person to decide where they want to wear their vital Identification.

E-Z-On Vest

Vests, harnesses and seat mounts for safe travel in school buses, the family car or emergency transport.

Medi-Pal

Seatbelt ID offers a quick and simple way to communicate vital health and contact information to First Responders.

Vacation and Travel info for Families with Special Needs Children

Most families with children choose to vacation and travel during the summer months, and it may take some planning to have a successful vacation. Even more so for families with children who have special needs.

If I Need Help

Identify your loved one with special needs with patches, shoe tags, ID cards, window clings, bumper stickers, dog tags, pins, clips and more.

AllerMates

Products to help kids with food allergies and asthma.

Angelman Family Contributions: Travel

Ask for bulkhead seating on airplanes. Helps prevent kicking someone in front!

Caitlyn

*** Southwest Airlines is especially accommodating!

When we fly on an airplane, we make sure to have our child with Angelman sit directly behind one of our other children or a parent. That way when he pulls hair, puts his hand through the seat, kicks the seat, it doesn't affect any other passengers.

Karen, wbk_sd@yahoo.com, angel Paul, age 15, Del+

My angel loves to go. If I say, "Let's go!" she is up and getting on her coat. Sometimes travel is difficult, but it's so worth it to see the look on her face when she gets to see something different and explore her world. I always keep a bag in my van with extra diapers, pants, shirts, snacks and other necessities.

Anne

We always carry a beach blanket that is waterproof when we visit friends and family. We put it on the floor or sofa to protect those areas in case she wets herself.

Prash and Mahendra, angel Lavania

Both my and my husband's family live a few states away, so travel is part of our life. Many years we have come home, and I've stated, "We are never doing that again!" But with experience, we have gained some wisdom and hopefully some helpful tips. 1. We don't eat in the car. This may sound counter-intuitive, but when we handed out a snack the fussing for more never ended. Bonus: the car is cleaner upon arrival. 2. We load the iPad with favorite movies and shows and attach this on the seat in front of our daughter. We have three other kids, so we put this where all can see but where our daughter cannot reach it. 3. So, yes, we do the iPad movie thing, but we try to start the trip with no electronics and instead listen to music or play car games. That way we can break out the electronics when things get crazy and kids get cranky. 4. We attach Fun and Function Chewy bite bands above my daughter's seat on that hanger rack thing. We can then attach her favorite chews and toys to them, so she can easily use them while we drive. 5. We duplicate her home sleeping arrangement wherever we go. We bring a mattress and Privacy Pop and a blanket from home. We've had some really hard nights when traveling, and this alone has helped the most.

Sarah, bnamommyisfun@yahoo.com, angel Lily, age 14, Del+

As hard as it can be to travel, the experience will lead to growth for your son or daughter. I have found that long trips, especially, will be followed later by a demonstrated spurt of learning.

Susan, angels Andreas and Yuan

Making the accommodations as similar to home as possible is key when traveling.

Shayna

When staying at a hotel we ask for an outside room. That way if we do not sleep we are less likely to keep anyone else awake. We are always upfront when making reservations and they try to keep the room on the inside empty too as long as the hotel is not full.

Becky

Whether your angel is in a wheelchair or not, we always request a wheelchair and a porter if available at the airport. It makes the security line so much easier and faster. It also frees up a hand or two.

Robin

A portable transport chair is beneficial during outings to amusement parks and malls but especially helpful while flying. (Even when your child is ambulatory.) It is easier to maneuver through airports. Also, airlines let you board early and change your seats to the bulkheads.

Janet and Rick, angel Chloe

We get the bulk seats. It provides extra room so our angel doesn't feel so confined. We bring along a DVD player, too. Our best vacation was to Disney Orlando our angel loved all the rides!!

Brisia, angel Mario, age 9

A tent is a great way to help travel with an Angel go smoother! We have set one up in hotel rooms, RV's and sailboats!

Alisa, alisa_giulietti@hotmail.com, angel Nicolas, UBE3A

U

UBE3A (See Causes of Angelman syndrome)

UBE3A mutations (11% of cases) – In these individuals, mutations in the UBE3A gene prevent its expression or function. Thus these individuals do not have the appropriate levels of functional UBE3A in the brain. (50% recurrence risk)

Angelman Family Contributions: UBE3A

When your child is diagnosed with AS, and if the child has the UBE3A mutation, make sure the mother is tested to see if she is a carrier. (That is, if you want to have more children.) That way you will know if there is a chance your next child could have AS.

AS Family Member

UNIPARENTAL DISOMY (See Causes of Angelman syndrome)

(UPD; 7% of cases) – In UPD cases, the individual has two copies of paternal Chromosome 15. Because UBE3A is not expressed from the paternal copy, these individuals lack normal levels of UBE3A in the brain. (Less than 1% recurrence risk)

V

VISUAL MEMORY

See E-Eye Issues

Angelman Family Contributions: Visual Memory

When our daughter was three years old she began taking a bus to school. One day, the bus had to take a different route home. The driver told us that our daughter began wailing and seemed to know that the driver was going the wrong way! After a similar incident happened again, we realized this wasn't a coincidence. She had remembered/memorized everything along the route! I believe this is common in our angels and it should teach us that this visual modality is vital. It, no doubt, offers them comfort in routines and stability. It also seems that our angel has great facial recognition, too!

Alice and Mark, angel Whitney, age 38, Del+ Class 1

VITAMINS

Angelman Family Contributions: Vitamins

Vitafusion Fiberwell Gummies Can be purchased at pharmacies, as well as Walmart, Target, etc.

This chewable supplement provides 5 grams of fiber; has a peach, strawberry and blackberry flavor; and can be cut in half for easier chewing.

Vitafusion Multi-Vitamin Gummies (Can be purchased at pharmacies, as well as Walmart, Target, etc.)

This fruit-flavored chewable supplement can be cut in half for easier chewing.)

<https://naturalvitality.com/natural-calm/>

Magnesium supplement that offers stress relief.

W

Walk

ASF Annual Fundraiser Walk



Since the 1999 first inaugural **ASF Walk** in Naperville, Illinois, this annual ASF event has grown to include nearly fifty cities across North America, including Canada and Mexico, with nearly 10,000 participants.

<https://www.angelman.org/walk/>

WALKING

Angelman Family Contributions: Walking

We had great success in teaching our grandson to walk in the swimming pool where the water helped keep him on his feet and legs.

Hardy, grandfather to angel Finn

Our daughter took almost five years to walk. Initially, we were told she would never walk. We insisted and provided her with Physical Therapy sessions during all this and she was literally taught how to crawl, stand up, and walk.

Harold

I wish someone had suggested we get a helmet for our daughter when she started to walk. Everyone was afraid she wouldn't like the helmet because of her sensory issues, but she loves it! Our PT recommended we try a front walker (instead of a rear walker) and that worked great.

Christine and Giovanni, angel Chiara

Our son started crawling when we started The Philadelphia Method best known as The Doman Method that helped him start the walking process. After creeping, came crawling and then the walking. It took about 3 years for him to learn how to walk. (Aged 2 to 5)

Brisia, México, angel Mario

When our angel was learning to walk and felt like he needed us to hold onto, we started putting a hula hoop around him so he could hold the front and we could hold the back to help support and guide him but he learned he didn't need our hands and fingers but he could feel "safe" with something to hold, plus it seemed fun! This helped him branch out and try on his own!

Andrea, mcneilak98@gmail.com, angel Tyler, age 18, Del+ Class 1

WEBPAGES AND SOCIAL MEDIA LINKS: ASF

ASF: www.angelman.org

Facebook: <https://www.facebook.com/AngelmanSyndromeFoundation>

Twitter: <https://twitter.com/angelman>

Instagram: https://www.instagram.com/angelman_asf/

Pinterest: <https://www.pinterest.com/angelmansyndfdn/>

YouTube: <https://www.youtube.com/user/AngelmanSyndromeFdn>

WEIGHT GAIN

Overweight / Obesity

- Present in 32% of individuals with AS, with obesity disproportionately affecting women

Angelman Family Contributions: Weight Gain

We try to feed our angel more natural foods. He has three meals and two snacks a day.

Brisia, angel Mario

WHAT ANGELS SAY TO THEIR MOMS

Thanks for never giving up on me. Your comfort and support help me take small steps forward that you might not even see. But, I AM moving forward and I AM truly learning every single day.

Thanks for searching deep inside me for parts that want to emerge. Mom, it is so exciting when a new connection is made between us.

Thanks for being patient when I am having a meltdown or when I am being stubborn. Sometimes I get really frustrated, frightened, anxious or overwhelmed.

Thanks for holding your head up high and being proud of me.

Thanks for working so hard, and I want you to know that I really do appreciate every little thing.

Thanks for telling me you love me. I really do understand. When I give you a hug, I am telling you that I love you more.

I love you to the moon and back, Mom, and I always will.

WHEELCHAIRS

One popular brand: Quickie <https://www.quickie-wheelchairs.com/>

Wheelchairs can be covered under your child's insurance plan. The physician can write a prescription and then you will be contacted by a vendor.

***** Transport Chairs are very lightweight and are great for short distance (airport, malls, etc.).**

https://www.amazon.com/s?k=transport+chairs&hvadid=78546404491535&hvbm=be&hvdev=c&hvqmt=e&tag=mh0b-20&ref=pd_sl_5q4kbcvq51_e

“WHO’S WHO” of Angelman syndrome

Please see the “S” Scientific Advisory Committee

<https://www.angelman.org/about/scientific-advisory-committee/>

Please see “C” Communication Advisory Committee

<https://www.angelman.org/about/communication-advisory-committee/>

Please visit the ASF website for a list of ASF-funded researchers.

<https://www.angelman.org/resources-education/resources/r/>

Also, visit the *ASF Funded Research 1996-Present*

<https://www.angelman.org/research/asf-funded-research/>

“WHO’S WHO” of the Angelman Syndrome Foundation

<https://www.angelman.org/about/>

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