Get Informed FAST

Newsletter for the Foundation for Angelman Syndrome Therapeutics

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Shooting for The Stars: Real Talk and Shining Examples in the World of Angelman Syndrome.

Dear Friends,

Already it is approaching the end of summer, and FAST has made some amazing achievements in the last few months, including two major fundraisers, plans for more, and helping to secure NIH funding for the Rare Disease Clinical Research Sites, allowing Angelman Syndrome research to continue! Additionally, FAST has had an expert team diligently working on future growth, and FAST is pleased to announce the culmination of efforts resulted in an absolutely stellar Business Plan that will guide the organization to amazing places in the near future.

In keeping with FAST's positive values, this issue is dedicated to the idea of hope. Hope, like a child, is a precious thing, and should be nurtured. When parents first are told of a diagnosis of Angelman Syndrome, it is natural to wonder what the future might hold. What does it mean to have a child with Angelman Syndrome? What will life be like? What are the right questions to ask, and of whom should they be asked? See our article on Variability in Angelman Syndrome, the first article in a series on the topic, to help address these questions.

What can you take away from this issue? These key points:

1. FAST is committed to a positive value set and is committed to finding viable treatments and therapies, made possible by an aggressive funding agenda.

2. No matter what, your child is a precious life, and through understanding the biology of variability in AS, you can nurture that life and dare to dream.

3. Great things happen every day, and FAST is committed to sharing the wonderful examples of daily living, and great experiences, had by Angelman Syndrome families and people.

It is okay to have Hope, to have High Expectations, and to pursue the very best quality of life your child is able to experience.

Shooting for The Stars,
Elke Sprow
Vice-Chair, FAST

The Foundation for Angelman Syndrome Therapeutics is pleased to announce the 2nd Annual FAST Gala, an "Evening With The Stars To Benefit The Angels", a black tie* fundraiser for Angelman Syndrome research. Our Guest of Honor this year is FAST friend and supporter, actor and Golden Globe winner, Colin Farrell. We are delighted to include Regie Hamm, winner of the 2008 American Idol Songwriter competition who will perform his hit, "Time of My Life", and the Chicago Symphony Chorus group "Sounds of the Season" as honored guests.

Sponsored in part by FAST corporate sponsors LexisNexis and Innovative Mag-Drive, this year's event will take place on Saturday, December 5, 2009 in the elegant Crystal Ballroom, located in the West Tower of the Hyatt Regency Chicago, 151 East Wacker Drive, Chicago, Illinois. The Crystal Ballroom is the only street level ballroom in Chicago, providing for spectacular

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views of the city lights and downtown landmarks.

The festivities will commence at 6:00 p.m. with cocktail hour, hors d'oeuvres and entertainment provided by both Regie Hamm on the piano and the Chicago Symphony Chorus performing a cappella holiday favorites.

A three-course dinner, with table-side choice of entrée, will follow the cocktail hour. There will be a presentation by Dr. Edwin Weeber highlighting the latest breakthroughs in Angelman Syndrome research. Dr. Weeber's research has received the prestigious distinction of being selected as the Scientific Highlight of the Year by the Federation of European Neuroscience Societies. Following Dr. Weeber, be prepared to dance the night away with help from Regie Hamm and the amazing band!

This year's extensive silent auction will provide fantastic holiday shopping opportunities with a vast variety of unique and interesting items, so don't forget to bring your wallet!We are thrilled to offer our guests a phenomenal room rate of just $99.00 per night. To book your reservation at the Hyatt Regency, click here. All of our guests can enjoy valet parking at just $10.00.

We look forward to seeing all of our returning friends this year as well as welcoming many, many new ones to this magical evening.

*Black tie optional

Cheers,

Paula Evans
Chairperson

Variability in Angelman Syndrome

What Does This Mean, and Why Does This Matter?

An important question that may arise with the diagnosis of Angelman Syndrome, and that may persist is "what will my child be like?". This is a very natural question following a diagnosis as families and caregivers seek to understand the implications of the diagnosis.

This article will address some basic scientific concepts upon which variability in genetic syndromes is grounded, as well as discuss the effects that the ideas and realities of genetic variability actually have for both the child and the family living with Angelman Syndrome (often referred to as "AS").

What Will My Child Be Like?

You may have noticed that your child is not achieving expected milestones and sought help with a medical professional. Due to the fact that it is a rare disorder, it is common for doctors to not at first suspect or test for Angelman Syndrome. As such, a diagnosis is sometimes not forthcoming. However, some diagnoses of Angelman Syndrome are made before one year of age.

When a parent is told the diagnosis is Angelman Syndrome and they are made familiar with the characteristics of this disorder, such as seizures, sleep problems, non-verbal communication, severe cognitive impairment and more, it is quite natural to wonder what the future may hold.

Early diagnosis can be beneficial. First, one has more time to come to terms with what it means to raise a child with special needs. This can be intimidating for there are many factors to consider including the following: special education; doctors and specialists; daily living; and long term living options. An early diagnosis also means an early start of therapies and access to services, which has been shown to be of enormous benefit to the child. A late diagnosis is equally important. While the parents/caregivers may already suspect that their child has special needs, getting a genetic diagnosis of AS opens the door to a wide base of knowledge and strategies that are effective in AS, as well as providing access to a community of others with shared experiences.

What will your child be like? It is a natural question to wonder if they will be able to sit up or walk. You may ask, "Will my child talk or use sign language? How will they play with other children? Will my child go to school? Will he/she be able to ride a bike? How will my child's development progress and how does that affect plans for the future?"

While no one can predict milestone attainment, we do know that not all children with AS experience the same degree and severity of symptoms. For example, while most children with AS will experience some form of seizure activity, some children with AS never have seizures.

In order to understand the manner in which these differences occur, we need to explore additional aspects affecting the genetics of Angelman Syndrome. (Please visit the Genetics page on the FAST website for an in-depth review of the basic genetics of AS). Two key concepts that can help explain variation in Angelman Syndrome are genetic penetrance and genetic expression.

Genetic Penetrance

Penetrance is a term used in genetics to explain the likelihood that a gene (genotype) will express an associated trait or appearance (phenotype). Populations expressing a genetic variation (like the loss of UBE3A function seen in Angelman Syndrome) are described by associating that genotype with certain characteristics. In Angelman Syndrome, the characteristics

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define the syndrome. For example, 70% of deletion children will have a sleep disorder, or 1% of all AS individuals will have mosaicism.

Penetrance can be described as:

1. Complete penetrance - all individuals having the gene variation have clinical symptoms associated with the variation.

2. Highly penetrant- the trait caused by the gene variation is almost always apparent.

3. Reduced penetrance - some individuals may not express the trait, although they carry the gene variation.

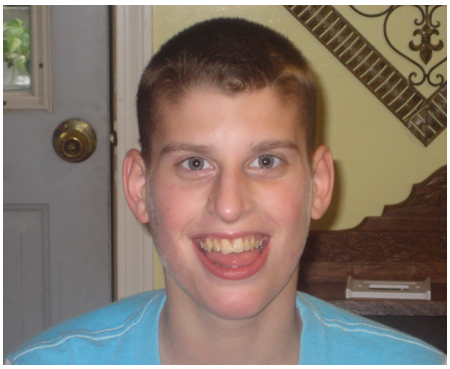
4. Low penetrance - a trait will rarely be apparent even when the gene variation exists. In this case it is often difficult to differentiate between genetic and environmental factors as a 'cause' of a trait.

The current body of research suggests that Angelman Syndrome falls into the realm of complete penetrance. That is, anyone with a mutation that affects maternal *UBE3A* expression will present with characteristics of Angelman Syndrome. However, there are different expressions of the phenotype among individuals. If we take into account the other 99.9% of inherited genes, as well as differentiated environmental influences, it is quite normal to find differences in abilities and development among those diagnosed with Angelman Syndrome.

Genetic Expression

Angelman Syndrome is just that....a syndrome. If developmental milestones and abilities are plotted, a normal bell shaped curve of distribution occurs. Some individuals will have minimal self-help abilities and need maximum care, while others may have more developed self-help skills and need less assistance.

While there is a recognized relationship between genotype and the severity of phenotype in Angelman Syndrome, the correlation is not definite. For example, there may be two children with Angelman Syndrome that share the same class of deletion size, yet there are manifest differences in milestone attainment and expression of abilities.



The following concepts can help explain this:

1. In Angelman Syndrome, there is a functional loss of the maternal *UBE3A* gene. When the maternal copy of the gene is nonfunctional, (or non expressive), Angelman Syndrome results. Without that functional copy of the gene, a myriad of problems occur, which among others results in a brain that cannot process experiences correctly, or lay down memories in typical fashion. This translates into cognitive delay, non verbal behavior, seizures, and more. The paternal *UBE3A* gene is still present, but remains 'silent', which is the normal state of the paternal gene.

2. Generally, children that are born with a deletion of the *UBE3A* region (encompassing more genes than just the *UBE3A* gene), tend to have greater challenges in terms of motor development and seizure control. The reason that deletion positive children are generally more severely affected is likely due to the loss of expression of many other genes in the region. Other genotypes of AS (UPD, ICD, *UBE3A* mutation) show a tendency to be more mildly impacted than deletion positive types, again likely due to the fact that people with these genotypes have not lost additional genes in the Angelman region of chromosome 15.

3. Environmental influences also play a large part in determining the developmental progress of children born with AS. Getting timely medical care and follow-up, experiencing physical, occupational and speech therapy and having a firm plan for schooling that supports the particular needs of the child can account for some variations in development, all other factors aside.

The message here is that your child is both the product of their ~29,000 other normally functioning genes, as well as their life experiences. A child who is diagnosed with Angelman Syndrome due to a genetic deletion may also look very different in terms of characteristics than those diagnosed with a non-deletion error (such as uniparental disomy), or an *UBE3A* mutation, or some type of mosaicism.

However, the genotype is not alone in determining the phenotype. While intense therapies such as those provided by Early Intervention Programs are normally directed toward children from birth to three, obtaining resources and providing therapies shown to be effective in Angelman Syndrome are important even if a child is diagnosed later in life. Effective therapies, and positive environmental influences, have been shown to increase developmental abilities, as well as enhance the quality of life for many individuals with AS. Medical professionals and local human services agencies can provide guidance toward resources that result in meaningful developmental enhancement.

While there is no present 'cure' for the genetic defect in Angelman Syndrome, it has been shown that physical, occupational, and speech therapies are beneficial for children with developmental delay, regardless of genotype or present developmental attainment. (For more information on these types of interventions, visit the Get Informed section of the FAST website.)

How Genetic Variation in Angelman Syndrome affects Your Child

As discussed above, there are variations in the presentation of Angelman Syndrome from individual to individual due to the combination of genetics and environment. As a parent or caregiver of an individual with AS, it is important to remember that nothing is set in stone. It is a mistake to presume that all of the characteristics associated with this syndrome will present in each individual to the degree described by clinicians for the syndrome in general.

In assessing developmental achievement or progress in such areas as "when did the child start to sit up, walk, potty train, play games, etc...", the variance is great, even among AS children of similar genotype.

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There are a few key factors to keep in mind that impact developmental attainment as well as "phenotype", or appearance:

Genetic Penetrance and Expression: While Angelman Syndrome may fall into the realm of complete penetrance, the expression of the other genes in the genome play strong roles in your child's abilities and personality.

Epigenetic Regulation: Outside influences (environment, diet, medications, etc…) may influence the level of skills a person will have through enhancing inherent potentials and managing developmentally disabling medical conditions with medical intervention.

Seizure Disorder: This can have a significant impact on the development of an individual and should be closely monitored by a licensed medical professional. Medications and dietary modifications have been shown to alleviate epilepsy and should be discussed with the child's licensed medical professional.

Therapy and Services: There is ample scientific evidence that points to the success of Early Intervention in terms of physical, occupational, and speech therapy for persons with disabilities. Therapies for older individuals can also be effective in enhancing skills and the quality of life.

Diet and Nutrition: Like any individual, a person with Angelman Syndrome needs a nutritious diet. Given the feeding and reflux issues many individuals with AS deal with, it is beneficial to seek advice from dieticians and GI specialists If a child is unable to take foods orally, parents should consult with a medical professional about a G-tube, which delivers nutrients through a surgically manufactured opening in the stomach.

Establish a 'circle of support' for the individual with Angelman Syndrome as well as for the parents/caregivers. This can include caregivers, friends, caseworkers, teachers, medical professionals, therapists and family members. The idea is to build a support network for the individual and parents/caregivers, so that as the individual grows, there are multiple resources to draw upon when needed. For example, it could mean something as simple as needing good respite care while the primary caregiver does household errands, or something as complex as arranging for special care when traveling. Also, having people around the individual that care about his/her welfare and growth, will go a long way toward allowing the individual to achieve his/her fullest potential through early childhood, the teenage years, and into adulthood.

A final thought to impart, is that while a person may be born with a loss of function of the one gene, *UBE3A* (the "Angelman" gene), she or he is, however, born with thousands of other fully functional genes. This, more than anything else, will determine "what your child will look like". Yes, an Angelman Syndrome diagnosis may be life-changing, but many other factors will influence development and determine the overall quality of life experience. Foremost are the variety of enriched environment experiences that allow for learning, growth and achievement.

The best thing any parent can do, is to get informed. Find a great doctor. Seek out other parents and join an Angelman Syndrome organization that best meets your values. Become an advocate and aim high. Small miracles happen every day. It makes good sense, to leave the door open to experience them.

(All of the individuals pictured in this article have Angelman Syndrome and are deletion positive.)

Article written by Elke Sprow.

Reviewed for accuracy by **Lynn M. Bird** MD Rady's Children's Hospital, San Diego, CA and **John R. Waterson** MD PhD FACMG Oakland Children's Hospital, Oakland, CA

Across the Syndrome - Profiles of Individuals Living With AS

Profile of Simon C.

Profile Date: 2009 at the time of the profile, Simon was 18 years old.

Filled Out By: His mom.

3 people he loves most in the world
He most loves his mom, dad, Aunt Sally, some school staff and camp counselors.

3 activities he loves most
He loves to attend live concerts and plays, (particularly if there is music), dance; going to parties; watching movies and certain cartoons, and swimming. He loves interactive toys, books and horses.

Does he have siblings? What are their ages?
No.

Describe his personality as an infant.
As an infant, Simon was very sweet and smiley, rarely cried, extremely enthusiastic and loving/super hyper - always fell asleep in the middle of playing, sometimes with hand still in air.

Has his personality changed over time?
Over time, he has more worries and more frustrations, so he shows his serious side more, but he's still very loving and sweet.

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He is still a very determined communicator, using many strategies. Simon is such a lovely person, interested in many things, a real people-lover, quite smart in many ways. His greatest fear is to be excluded.

At what age did he sit up? Crawl? Walk?
He missed early major milestones but walks very well now. He sat up around 7 months, crawled before that, and could pull to a stand and cruise for a couple years before he finally walked alone, at the age of 3. He's a fast runner, can walk backwards well, can walk for miles, and is pretty good on difficult terrains. His balance has improved immeasurably since he was young. We do nature obstacle courses with him, over river stones, over fallen trees, stepping over narrow streams, going under low boughs - he loves it.

How does he prefer to communicate?
He had a definite "lost words syndrome" and spoke around 30 word/word approximations. Many were never repeated, several hung on a while, and the last one "mama" disappeared around age 11. He is able to say it occasionally. He has his own signs, we were steered away from sign language for him, which I believe was a mistake. He is quite adept with PECS and other picture systems, with and without voice output. He has a dynamic screen device that is not very easy for him to access, however, he is able to go through several screens to get what he wants if he's motivated enough. We are in search of something better. Building a comprehensive communication system is a daunting task, but there is none more important.

When other methods aren't available, he sometimes still grabs,. When he's calm and isn't anxious about not being able to communicate, he's such a delight and can be very polite about asking. He's very good at eye contact, leading and bringing you actual items in case you didn't catch the request another way.

Has he ever used a communication device? What kind? Is it meaningful to him? Does he use it without prompting?
He has used several. At home, for the dinner table, he uses a GoTalk 9. He uses it without prompting, and will use his icons also without prompting. If he needs prompting, he gets back on track quickly. He had a Chat PC 2, (on a pocket PC platform) which is a nice little thing but for his fine motor skill level, it was never a good match. It also broke frequently. We are trialing some DynaVox devices starting next week, through his school.

Which therapy has he most loved? What made it fun?
He used to love swimming above all, then later became more interested in riding horses. He still loves horses but will sometimes refuse to ride, which I connect with his surgery for turned-in foot and later a broken knee. We will be trying again this year. He has always enjoyed communication therapy if he had a good therapist. Some have been wonderfully creative, inventing interactive computer games just for his likes and motor skills, and have had him asking for 'real" activities as well, so that he could ask electronically, and also give commands for what the other person would do. Again, if he's had a good OT, he's enjoyed that - learning to unscrew bottles, turn on the water and wash his hands or orient the cup under the faucet for a drink and other practical things he obviously wanted to learn. He loved water therapy after his broken knee last year. Can't think of any others right now, but want to make the point that he enjoys learning and enjoys people, so if you have the right people teaching the right things - he makes fantastic progress and loves it.

Which therapy has he liked the least? What made it bad?
Again, it's the people. If he feels forced, or bored, or the person doesn't tune in to him, he will not like anything. He doesn't do things just because you ask him to. He will try just about anything, so you have a chance to teach him many things. Making it interesting is the challenge of the therapist or teacher.

Does he attend school or a daily program?
Simon goes to school 5 days a week. He leaves for school on the bus at 8:30 and returns home at 3:00 pm.

Does he enjoy school/his daily program? What does he enjoy most? What does he enjoy least?
He loves school because he loves the people. Staff turnover is difficult for him, because he misses people, but he has no trouble accepting new people. They have a community outing every day, he likes that, he loves the weekly swim at the YMCA, he likes this speech therapist and her classes. He likes variety in the lessons. I found out they were using the same pictures for over 6 months in one class, and he was so bored he was acting out. He likes routines, but with changing materials. He also doesn't like being pushed around - some people working with DD people think they have that right. But with Simon it really backfires on them.

Is he involved in any extracurricular activities?
He was involved with KEENusa for a couple of years, but it's very limited here (smallest space of any KEEN program) centered around a small gym, so he lost interest. Simon goes to camp every year, this year he went to sleepaway camps in June, July and August. (overnight camps that last for 4 to 6 days). He truly loves to go to camp, it's freeing for him and he loves the activities and the counselors. We go to a yearly 3-day family camp, (for families with disabilities) which he also loves. This is a camp that recruits college students to volunteer for the weekend, they have scheduled activities for the kids while the parents go to classes, rest and socialize. There is also family time, to swim or do other activities, boating, walks, etc. All meals are provided in the community center, and it's a great family experience. We've been going to that camp for 7 years now.

In the past, he's participated in Little League (challenger team) and AYSO soccer (they have a special needs group). He much preferred the baseball, which is more of a turn-taking sport. With soccer, he didn't like all the kids running around him. He also has had regular

horseback riding for several years, but had to drop out due to surgery and a broken knee (2006 and 2007 respectively). He's also been involved with Special Olympics aquatics but they had issues with
not enough volunteers so he quit. He is now not so excited about cold water pools

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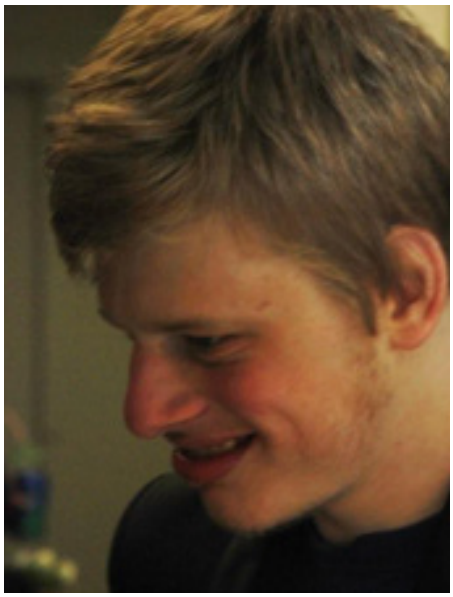
unless it's warm weather. He prefers his therapeutic swim class in the warm pool.

After school, we do walks or visit kid-friendly museums or zoo in summer. In winter it gets dark early, so he's limited in outside activities. He loves his TV a little too much.
There isn't much social opportunity here after school, and I'm working hard at creating an after school program thru ARC.



Where does he live?
We have built a separate apartment for him attached to our house, and Simon is in a partial supported living situation. He sleeps in his apartment 5 nights a week, and eats dinner there 3 nights a week. The rest of the time he's home with us. Finding full time staff has been difficult.

What is a typical night of sleep like for him?
He generally sleeps through the night. If he needs to go to the bathroom he may get up and ask for help, but goes back to bed right away. We find melatonin essential – 1 mg. sublingual at bedtime.

Does he have seizures? What age did they appear? Are they controlled with medications?
The seizures were very bad pre-puberty. They snowballed from petit mals to several types of partial and generalized seizures. We luckily found the right drug (felbamate - a low dose) which controlled through that period. Now the epilepsy is not a big issue. He has occasional petit mals, which correlate with being ill or having an infection. He hasn't had a grand mal seizure in several years. He also takes clonazapam, since age 2.

What do the seizures look like? Have they changed over time?
At age 2 he received the epilepsy diagnosis, was ill with fever and had 2 grand mals and was hospitalized. He was put on klonopin (after trying several other things that exacerbated things). He was prone to staring spells, the major type. Later, around 8, he started having more and longer seizures and was put on Depakote. This made everything so much worse and the types of seizures proliferated until he got a diagnosis of Lennox Gastaut Syndrome. He had at least 7 types of seizures, culminating in total drops. Just as we were getting him a helmet, we switched neurologists and he went on Lamictal. That helped, but only a few months. Then the doc convinced me to get him on felbamate, and that truly saved his life. Even though it's been about 9 years, his dose has decreased and is quite small.

What medications is he on and why?
Clonazapam and felbamate for seizures.

Does he have any medical issues related to AS such as reflux, g-tube, etc.?
He had fairly bad reflux until he walked, at age 3, exactly as his pediatrician predicted. Seizures. Difficulty controlling body temps in extreme weather conditions.
Cavus foot that required surgery to straighten.

Does he have any medical issues that are probably not related to AS such as asthma, allergies, etc.?
He gets intermittent eczema or psoriasis, mostly on his feet and head. Fish oil supplements are very helpful for that. He had an orchiopexy around age 8.

What is your child's genotype? When was he diagnosed?
Deletion positive. Formally diagnosed at age 10 via methylation test, then the FISH test confirming the deletion was done one year later.

Please share a piece of advice that was most helpful to you regarding your child.
Don't hold dad to your standards - it's ok for dad to hang around and watch football with him sometimes, and just do dad things. Most dads are not as detail-oriented as mothers. Every moment doesn't have to be a teaching moment. Your child mostly just wants you to be present, and be yourself and not always be caught up in the roles of teacher/therapist. Your child doesn't want to be fixed – he just wants you to love him.

What is the most important thing you wish others knew about your child?
He's a treasure just as he is. He is smart and doesn't want to be talked to like a baby. He has his own strengths and weaknesses, like everyone. He wants to be accepted. He doesn't want to be pushed around or counted out.

If there is one thing you wished someone had told you earlier that you would like to share, what would it be?
This I get asked constantly by parents with younger angels.

My answer is:
1) Focus on daily living skills EARLY, don't wait until the child is big to teach him how to pull up/down his pants or how to climb into the car seat. PT and OT should be about helping your child learn to do very practical things for himself.

2) Focus on communication always. Communication using icons should always be labeled with words, as this is a way to teach reading sight words. I believe reading is the most important academic skill. Teach conversation and not just "I want this, I want that."

3) Teach social skills. Use the communication for social commenting and feelings as well as requesting items and activities. Nuture friendships very early, friends are precious and hard to come by as children with disabilities age.

These are the 3 biggest issues as your child grows, yet in IEPs they can really waste your child's time with non-essentials.

Do not be lenient when they are bratty. Do not give things because you feel sorry for the child. Teach manners, like asking appropriately. (Do not reward headlocks, biting, and other obnoxious behaviors and don't let anyone else! What's cute as a baby is a huge crisis when they're big and strong). Provide communication methods, try everything.

Teach to their interests and don't force your curriculum if it's not tailored to their interests. Feed the kid well! Don't wait until they're 2 to introduce vegetables. Kids like familiar foods, make healthy foods familiar from the beginning. Don't do the sugar thing - besides health problems, taking care of their teeth can be a big, big challenge.

Profile of Delaney VB

Profile Date: 2009 - At the time of the profile, Delaney was seven years old.

Filled Out By: Her mom.

3 people she loves most in the world
It's hard to pick just three people! Delaney LOVES her Grandparents, Aunt and Cousin! Of course she also LOVES her Mom, Dad, and brother Sean, but then again, I think that Delaney truly LOVES everyone she meets!!

3 activities she loves most
Delaney loves to swim, watch movies (Barney, Disney Sing Alongs) and doing "school work" (sitting at the table and coloring, doodling with a pencil or pen, looking at flashcards, etc.) play outside, ride her Skitter Toy, play ball

3 things she loves most
Delaney loves Little Debbie snacks, school, and going shopping with Mom and waving at everyone who goes by – everyone is a "friend"!

Does she have siblings? What are their ages?
Delaney has an older brother, Sean, who is 13 years old. I know they are not siblings, but Delaney thinks of her dog, Buddy, as her big brother also.

Describe her personality as an infant
Delaney was always a very calm/mellow infant, but she was extremely fussy from around 7pm – 10 pm every night between 1 week old to 6 weeks old. At 6 weeks of age, Delaney was hospitalized for "failure to thrive" and had low blood sugar problems. She was hooked up to all kinds of wires, machines, etc, so it was really hard to hold her much and she definitely became a much easier baby after that. I think that this is because she learned to soothe and calm herself while in the hospital.

Has her personality changed over time?
Delaney is still pretty laid back as a child, but very "busy". She is very active and gets into more things as she is walking and running and can now reach almost any cabinet in our house. She is pretty good about knowing and respecting her limits and really has not been much of a problem as to safety and security in the home. She is pretty conservative and respects and stays away from situations that are dangerous. I do think that her attention span is getting better as she gets older though!

At what age did she sit up? Crawl? Walk?
Delaney walks and/or runs everywhere! She sat up by herself if you placed her in the sitting position at 8 months of age and she was able to get to the sitting position independently at 13 ½ months. She commando crawled at 16 ½ months and then crawled on all fours at 20 months. Delaney pushed a toy shopping cart to walk with at 21 months and then walked independently at 26 months – a month prior to her AS diagnosis so we have always felt that she set the bar/standard very high for herself!

How does she prefer to communicate?
Currently, Delaney uses a DynaVox MT4 for communication. It has 16 buttons available to her on each "page" and she probably has over 100 pages for her to access (and she uses them all) at this time! Delaney also uses a lot of gestures, about 15-20 signs, and two words to communicate with (mama and bubba). Delaney said her first word "mama" at 4 years old. She then said "bubba – for her brother Sean and for the dog Buddy" at 4.2 years of age. She has made various sounds over time such as the mmmmm sound for more, but not consistently. She also uses the mmmm sound for singing and will hum along with a song.

Has s/he ever used a communication device? What kind? Is it meaningful to her, does she use it without prompting?
Delaney has used a number of different devices for communication over the years. She started using a HipTalker at age two. This worked well for her at that time and was great for socialization. It had 4 buttons on it and she used it pretty functionally for greetings, farewells and to ask "Can I play?" for example. Once Delaney started attending Scottish Rites Preschool (Speech and Language program which focuses on AAC) they started her on a 7 level Communication Builder. Again, this worked well and it had 8 buttons for her to use, but it was very difficult changing out overlays throughout the day for her to use in specific settings – snack time, circle time, free play, etc. She used the Communication Builder from about 3 – 4 years of age. Right before Delaney turned 4, Scottish Rites introduced her to the DynaVox MT4. This was life changing for Delaney, our family and her school! I would have never dreamed that Delaney would be able to access and move from page to page trying to find what she wants to say, but she does! She will go through 4 or 5 screens or more sometimes to find what she wants to say!! Delaney uses the DynaVox throughout the school day. She uses it to communicate with her peers and teachers by asking questions, commenting and playing games. She also uses the device for "work" activities – matching or naming numbers, letters, colors, book talks, literacy, etc. We LOVE the DynaVox and the voice it gives her!

Which therapy has she most loved? What made it fun?
Delaney loves going to HippoTherapy, gymnastics class and dance class. Granted, these activities (other than HippoTherapy) are not "therapy" and I think that is why she likes them so much!!

Which therapy did she most dislike? What made it bad?
As an infant/toddler, Delaney hated physical therapy. She hated to be on her tummy and when the physical therapist would come to the house, I think she knew that the therapist would make her get on her tummy and she would choose to "shut down" and fall asleep. We spent a lot of time and money on "therapy" – OT, PT, and Speech – when Delaney was younger. It was so stressful getting her to and from the appointments and keeping up with Sean's activities, sports practices, etc. Delaney seemed to make the most progress and learn the most from her brother – he got her to sit up by herself, take her first steps and learn how to throw and kick a ball. Sean has been an exceptional big brother for Delaney and has had the determination and patience to help her excel – when he was much younger and was told that Delaney most likely would not talk, he replied that he would teach her! We figured out after a long time of doing therapy, that Delaney got just as much "therapy" by attending "typical" activities like her peers – dance, gymnastics, soccer, etc. She enjoyed these activities much more than she ever enjoyed therapy sessions and we saved a ton of money!!

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Does she enjoy school? What does she enjoy most? What does she least enjoy?
Delaney LOVES school – being with her peers, the socialization, and structure of the classroom. She has been fully included in a regular classroom for Kindergarten, first grade and now second grade. She is rarely pulled out of the regular classroom except for adaptive PE and sometimes extra work with the speech/path. She has a one-on-one paraprofessional (actually several that "share" Delaney), but the para has been instructed to not "hover" and Delaney is pretty independent during the school day. She knows the routine, follows directions well and is often the first to go to the assigned activity, line up for recess, lunch, etc. Probably having to exercise self-control is the one thing she dislikes – last year she had to go to the "think chair" for eating an M&M that they were using for a math/counting exercise – but what 6-year old wouldn't!!!.

Is she involved in any extracurricular activities?
Delaney attends a special needs dance and gymnastics class. She also attends Sunday school and church. She has played on a soccer team and is a number #1 fan at all her brother's baseball and football games. She went to a cheerleading clinic last year and "performed" with all the cheerleaders at a local high school football game.

Where does she live?
Delaney lives at home with us. My ideal living situation for Delaney in the future would be for us to buy a duplex/condo and Delaney would live with a "friend" and caregivers in one side of the house and then we would live in the other side. This way we could monitor what is going on with Delaney and be there to help out as needed, but she could have her independence and so could we.

What is a typical night of sleep like for him/her?
I'm embarrassed to admit that Delaney sleeps with me in my bed and that my husband sleeps in Delaney's bed. We have got to get this changed soon, but Delaney sleeps so much better when she is sleeping next to someone. If she is sleeping with me, she will only wake one or two times a night for a few minutes on most nights. It seems like once a month or twice a month maximum, Delaney will decide to have a party early in the morning. She will wake at 1 or 2am and not go back to sleep. I'm never sure if this is because she has a stomach ache, nightmare, or just what??

Does your child have seizures? What age did they appear? Are they controlled with medications? What do the seizures look like? Have they changed over time?
Delaney had an EEG done when she was first diagnosed with AS at 27 months. The EEG showed abnormal spikes and waves and a high likelihood for seizures. The doctor recommended we put her on meds and we declined. Delaney was a new walker at this time and we would notice times that she would stumble, lose her balance, grab the wall or furniture for balance, but at that time we attributed these instances to her newly acquired skill of walking – looking back we now realize that this was probably seizure activity. During these episodes she would quickly recover and always be off and on the go again. Delaney started Lamictal at age 3 years after some definite drop seizures. She still has drop seizures, startle seizures when her arms jerk and fly up in the air. She has also had strange eye blinking seizures. The seizures seem to be controlled by her medicine for the most part, she usually only has breakthrough seizures when she is ill with a sinus infection or UTI.

What medications is she on and why?
Delaney currently takes Lamicatal, Keppra (added at 5 years of age) and Topomax (added last January during a week-long hospital stay for seizure control). Delaney is almost weaned off the Topomax at this time. She also takes Vitamin B6 and an Omega 3 /Fish Oil supplement. Delaney takes Sal-Tropine to help reduce/control her drooling – she has been on this for 2 years now with no problem.

Does she have any medical issues related to AS such as reflux, g-tube, etc?
Delaney had severe reflux as an infant which probably caused the "failure to thrive" diagnosis at 6 weeks of age. She was fed by a NG tube while in the hospital for 2 weeks at 6 weeks of age and then 2 more weeks at home – what a nightmare – the doctors thought she was possibly suffering from a metabolic issue. She had low blood sugar and during one hospital visit had 80+ needle sticks in her heels – another nightmare for us all! Delaney is a very good eater now and is in the 80%-90% range for both height and weight!! Delaney also wears glasses for farsightedness and inserts in her shoes (hotdogs) to help keep her feet from rolling inward.

Does she have any medical issues that are probably not related to AS such as asthma, allergies etc.?
Delaney is pretty healthy overall! She does seem susceptible to UTI's and did have her tonsils and adenoids removed at age 3.

What is your child's genotype? When was she diagnosed?
Delaney is UBE3A. She was diagnosed at 27 months.

Please share a piece of advice that was most helpful to you regarding your child.
Since Delaney was not diagnosed until age 27 months, we had all that time of treating Delaney as a "normal" child and waiting for her to catch up and reach the missed milestones. When I refer to a "normal" child, I am referring to the fact that we took Delaney everywhere and treated her the same as any other child with the same expectations and goals. She attended an excellent in-home daycare and again the same expectations and behavior were expected from Delaney as the other children – to include self-feeding, napping on her own cot and staying on the cot until the lights were turned on, toileting, sitting in time-out if needed, etc.

What is the most important thing you wish others knew about your child?
Delaney is the constant "light" in our lives – her smile is dynamic, her laughter is contagious and her sense of humor is a delight to all who know and love her! Delaney understands everything that is going on in her environment and almost everything that is said to her. She is typically the first one to laugh at any joke or funny story that anyone tells!!

If there is one thing you wished someone had told you earlier that you would like to share, what would it be?
One thing I wish I would have done differently for Delaney is to have sent her to a "regular" preschool versus the school district's special education preschool. Delaney just didn't have very good peer models for language. We did only send her 3 days a week (it was a 5 day a week program) because she was already at a wonderful home daycare with all typical peers and I really wanted her to

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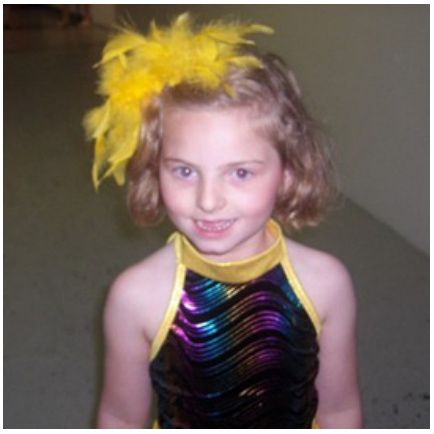
have as much exposure to typical children as possible. I felt also that the sped preschool with only 8 – 10 children did not prepare her for a Kindergarten classroom with 20+ students in it. It took her a while to adjust to learning how to interact appropriately with all the children and how to sit at the circle, etc.



Across the Syndrome is a new feature at FAST. Profiles are compiled by Erin Sheldon.

Do you know an individual you think should be featured in upcoming newsletters? If so, please contact us and we will send you our questionnaire.

Funding Approved For the Natural History Study

FAST is thrilled to report a very successful visit to the NIH which helped ensure another five years of funding for the Angelman Syndrome/Prader-Willi/Rett Syndrome Natural History Study! FAST, in a jointly arranged visit with Rett Syndrome International, met in May with the Directors of NICHD, including Dr. Duane Alexander and Dr. James Hanson, to discuss the critical need for the Natural History Study to continue even though funding had been cut for this program. In July, we were told that the two years of funding we had been cautioned to only hope for had been extended to the entire five years requested - a home run!

Our joint message of promising potential treatments currently under development that could be immediately trialed through the Natural History Study sites was well received. The NIH was pleased to hear from both an organizational and a parent perspective about the positive impacts that this NIH-funded study is having; we now are starting to acquire enough data to evaluate effects of various therapeutics, draw prospective conclusions on the genotype/phenotype relationships, and assist actual clinic al trials of promising therapeutics, such as levo-dopa. The related levo-dopa study was recently launched as a separate study using some of the same Natural History Study Sites to evaluate motor skill and cognitive improvements in Angelman Syndrome.

The NIH shared our excitement about the development of treatments through mechanisms and pathways that can be applied to two or more of these genetically similar conditions (AS/PWS/Rett) and potentially many other neurocognitive disorders. The NIH believes, and rightfully so, that it is getting the most for its' money by funding the joint Natural History Study at study sites across the US (Boston MA, Vanderbilt TN, Baylor TX, Greenwood SC, San Diego CA). As these sites are now well established, it becomes easier and cost-effective to start additional studies and trials using the personnel and infrastructure developed at these locations.

The NIH seemed to be most impressed with our consolidated advocacy approach in supporting the study. The NIH professionals are very dedicated and caring. One point that really hit home for me was when Dr. Hansen expressed that they "were here for us, no more importantly, here for your kids." In subsequent conversations with NIH representatives, we were told they received over 300 letters from the AS/PWS/Rett community in support of the continuing study and they expressed their appreciation to those who took the time to let them know how important this NIH-funded work is - so a big thank you to everyone out who voiced your support!

To learn more about the Rare Disease Clinical Research Network and/or to enroll in the Natural History Study, please click here

Article written by Sharon Claridge

Golfing For A Cure

The law firm of Koeller Nebeker Carlson & Haluck organizes a fundraiser for FAST

The weather forecast may have called for rain, but the golfer's prayer prevailed. The skies were clear, the sun was shining and the temperature could not have been more perfect for a day of golf. The law firm of Koeller Nebeker Carlson & Haluck's ("KNCH") first annual Golfing for a Cure Tournament benefiting the Foundation for Angelman Syndrome Therapeutics was off to a great start. The tournament setting at the Lincoln Hills Golf Club was picturesque with beautifully manicured fairways. The participants arrived at 11:30 a.m. on the morning of the event, anxious to hit the links.

Chad Dunigan, a partner with KNCH and a parent of a child with Angelman Syndrome ("AS"), organized the event and greeted the hundred golfers who then jumped into their golf carts for the shotgun start. Despite rainy weather prior to the

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event, the course played exceptionally well. As play ensued, the foursomes were greeted at each hole by corporate sponsors with various "giveaways" and as much unsolicited instruction as the players could tolerate.

Doug Tackett, Chad Dunigan, Dr. Edwin Weeber, and Michael Evans

Satiated with golfing, the crowd re-grouped at the Clubhouse for cocktails and hors d'ouevres. The food was as beautifully displayed, as it was delicious.

Avelar & Associates Group

The conversation was lively with tales of the game and there was plenty of social and professional networking amongst the crowd. Cocktail hour was followed by a lovely buffet dinner.

During dinner, Chad entertained the guests with a humorous recounting of the day's highlights. Gifts were awarded to the best performers and prizes distributed to a plentitude of winners. After Chad extended heartfelt gratitude for the support of his friends and colleagues, he turned the stage over to Dr. Edwin Weeber who managed to entertainingly educate the crowd about Angelman Syndrome and his recent work of curing this disorder in the mouse model. Dr. Weeber was as big a hit among non-AS parents as he has been among the AS community. After Dr. Weeber's presentation, the guests enjoyed a touching video presentation of children with AS,

Stacie Vetterli and Karryl Downing

which was created by Maiddy Dunigan. The night concluded with a silent auction where guests walked away with everything from sports memorabilia and premium wines to movie and massage certificates.

KNCH raised $28,970 for Angelman Syndrome research and the Foundation for Angelman Syndrome Therapeutics could not be more appreciative of their time, effort and passion towards our cause. We are already looking forward to next year's event and the fun and success it is sure to be. Stay tuned for announcements to next year's event!

Article written by Maiddy Dunigan

A Morning to Remember

Songwriter Regie Hamm Strikes a Chord with a Special Audience...

The sights and sounds of the big apple are intoxicating. New York City can truly make you feel like a conquering hero if you're doing something exciting there. I've been there more times than I can count, and I've had some absolutely cinematic moments in the city that never sleeps. I remember my first trip to the legendary metropolis. It was the pre-Giuliani era, and within my first hour in Times Square I had been held up at gun point and openly propositioned by a lady of the evening (wearing absolutely nothing under her fur coat) - all before I had gotten off 42nd street. I've been escorted out of a high rise, corner office overlooking central park, over a contract dispute. I once laid on the ground in Battery Park and watched the sun come up over the Twin Towers, with my wife, while eating bagels. I've heard grungy blues down in the village, I've been to shows and plays and tapings. I've performed in NYC many times as well, and it has always been a great experience. It's one of my favorite places on earth and it never fails to leave with an indelible mark of some kind.

Photo by Frank Koester/Spectrum Magazine

My recent trip there was no exception. After a whirlwind year as the American Idol songwriting champ in 2008, I had been fortunate enough to watch my song go from "Idol Finale Song", to "that moment" song, to a chart-topping hit that resided at number 1 on the pop Adult Contemporary chart for a record 16 weeks. The song, however, meant more to me and many in the "Angelman Syndrome" community, than just music business success. The "Full Circle" story behind it took on a life of it's own and brought interviews and blog readers and well-wishers from all over the world. It had been one of the most special and serendipitous years of my life. The culmination of 2008 was an awards ceremony in New York City (fittingly) on May 12th, 2009. The ceremony was all I could've dreamed of. Seated at Paul Shaffer's table for dinner along with my wife and two wonderful friends from the AS

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community, I enjoyed the music and awards being presented. Soon enough, I was on stage singing my signature song from that year and accepting the award for "Pop Song Of The Year". Once again, NYC had not let me down.

The rest of the week was a blur of shows and meetings and ended in a performance in the East Village that was my worst NYC performance ever. I felt a bit let down. I've never left the city on a sour note before. I didn't like it. The following day I would make an appearance at a place called the Rebecca School, a therapeutic day school for children with neurodevelopmental disorders, then be on a plane back to Nashville. Oh well, most of the trip was productive and great.

Photo by Frank Koester/Spectrum Magazine

The morning of the 15th was business as usual. Yolanda and I packed and met my manager outside the hotel at the car. An old friend of mine just happened to be in town and decided to ride along with us and see the school for himself. We arrived at the Rebecca school and figured we would have a good 45 minutes with the kids and then be on our way. Though I am raising a daughter with AS and am around special needs kids often, I wasn't sure what to expect. It's one thing to be daddy, it's quite another to be the "entertainment" for 60 plus kids with autism. I was nervous. Just before they brought the kids in, my wife gave me the tip of the week..."play Sesame Street - they'll love it!" When the kids were in and assembled, I stared at their beautiful, troubled faces. I recognized the facial tics and uncomfortable laughter and inappropriate movements all too well. I suddenly felt strangely at home and began doing my own version of the Carpenters' classic "Close To You." It seemed to settle the kids and at the end I received applause that made me breathe a sigh of relief. Then I dove into the suggestion of my brilliant wife and did my daughter's favorite song..."Sesame Street." By the end of the second verse I had a chorus

Photo by Frank Koester/Spectrum Magazine

Behind me yelling the answer to the perennial question, "can you tell me how to get ...how to get to ...SESAME STREET!!!" Sung at the top of their lungs. My big finish was my song sung by David Cook that has meant so much to so many. After the first chorus, I saw two children stand up and begin waltzing to the six eight rhythm. As they swayed from side to side, I caught their teacher out of the corner of my eye. She was sobbing uncontrollably. I took a quick glance behind me to see a room full of children with Autism (one AS child who had been mis-diagnosed), dancing and singing and hugging. The teachers and handlers were all in different stages of shedding tears. It was all I could do to choke back my own and get through the remainder of the morning.

I finished my song and immediately went to embrace all of those babies I could get my arms around. I was hugging the teachers too. Those 20 minutes had officially topped anything that had happened with that song all year. The reason for writing that song, or any song, was clear to me in looking back at crooked smiles and awkward motion. May the 15th 2009 was the best show I've ever done. Thank you Rebecca School. Thank you New York City. In the teary words of my friend, who witnessed the whole thing, "I will never forget this." Amen.

Article written by Regie Hamm

Angelman Syndrome Down Under

A Mothers Plea Brings AS Information, Education, and Awareness to Australia

It all started with a desperate plea from an Aussie mum... will you come to Australia; tell us first hand about your research and what it would mean for our kids? We never imagined that the ramifications would be so positive for Angelman Syndrome families in Australia.

Dr. Jessica Banko, Dr. Edwin Weeber and Dr. Honey Heussler

From the moment Dr. Weeber agreed to come to Australia it was a frenzy of emails between a few AS families working out a way for it to happen. The momentum was building, and truth be told, there were a lot of butterflies about whether we could make this happen. But due to the generous nature of some of our medical professionals and hospitals, we were able to provide families and clinicians some valueable time

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with Dr. Weeber.

Jen and Mario with daughter Lucy

Dr. Weeber was so generous giving his time to parents in Brisbane and Sydney and the sessions were a resounding success. The sessions included walking us through the genetics behind AS, creating a representative mouse model and the subsequent rescue of that mouse. Parents were astounded to hear the numerous ways in which Dr. Weeber's lab is searching for a therapeutic intervention for our kids. We could see first hand how this has all been developed.

Meagan and Rohan with daughter Molly

For some parents it was the first time they had heard of any research into AS. Only five years ago parents in Australia were told that there would never be any research into AS, and that they should take their children home and love them. This is the first opportunity for some parents to hold out any hope to hear the voices of their angels.

Dr. Weeber's sessions with the clinicians and scientists in both Brisbane and Sydney were again well attended. Our hope for these sessions was two-fold - that the clinicians would have a better understanding of AS for future diagnosis, and that scientists would take an interest in our children's disorder.

We truly believe that there would have been no other way to inject this level of excitement into the AS community in Australia. No amount of reports, or articles, would have matched giving parents the opportunity to meet and chat with Dr. Weeber. And we were truly lucky that Dr. Weeber's equally talented wife, Dr. Banko, came to visit us as well. It was almost like getting a two for one deal - two brilliantly dedicated and compassionate individuals who truly care for the future of our angels.

And the generosity continued with the Mater Children's Hospital in Brisbane offering to set up DNA sequencing for parents without a confirmed diagnosis. Previous to this tests for mutations were sent overseas generating long waits and huge expenses to parents. We also managed to gain some precious airtime with a national news network running a story on Meagan and her angel, Molly.

The feedback from parents has been incredible - one parent described the session as a life changing moment - at last they finally had some hope. Parents were able to understand why AS is so simple in its design, yet so devastating in its effects. Why there really is hope for a cure. We feel that there is now a reinvigorated AS community in Australia. It is our goal to build on this to increase awareness in the general public, to reduce misdiagnosis through providing important information to our medical community and to encourage research in Australia so we can all work together in the search for a cure.

And for all that Ed and Jessica brought to Australia (yes we were mates by the end of the first day formal titles were put aside), we hope that we have given some back to them. They now know that "Oz" isn't a town in Australia - it is just slang for Australia, that we sometimes raffle trays of meat in pubs (yes we do have butchers, but this is way more fun), and that after you have seen 100 kangaroos they even get a little boring!

Melanie with son Lachie

We could never convey just how appreciative we are to Ed and Jessica for taking the time to reinvigorate a nation of angel families. They, along with anyone else in the international AS community, are welcome at our shores whenever they feel the need to buy a ticket in a meat raffle.

Many thanks, Meagan Cross and Jen Kyriacou

Photos by Emily McInnes
