

Children with Ataxia



National Ataxia Foundation

Children with Ataxia

Table of Contents

Introduction	1
Alaysha's Story	2
How You Can Help	3
Angela's Story	4
Cameron's Story	6
Liesel's Story	7
Maya's Story	8
Olivia's Story	10
Handwritten Note	12
Yasin's Story	13
In Memory	14
NAF Is Here to Help	15
About NAF	16

Introduction

It is estimated that 150,000 individuals in the United States are affected by ataxia. Sadly, many of those who are affected are children.

Families who have a child with ataxia are forced to adapt to the constant change and uncertainty of this disease. The impact from the disease can be felt not only by the child, but by the family as a whole. There is struggle, helplessness, and heartbreak.

But there is also courage, hope, and a commitment to making the most out of *today*.

The stories presented in this booklet are the stories of children with ataxia. Parents and family members have generously provided these stories to share their difficult and emotional ataxia journey. They offer a candid perspective on how ataxia has forever changed their lives and on their hopes for the future.



Alaysha's Story

Story provided by her Mom

On February 4, 2015, Alaysha, my eight-year-old daughter, was diagnosed with ataxia telangiectasia (A-T). When she was a baby they tried to diagnose her but couldn't figure it out, so we left with a general diagnosis of ataxia. As she grew it even seemed like she was growing out of this wobbling. She met all her therapy goals, so we just went on living life until this school year.

Her walking and talking started getting worse so I felt it was time to go to the neurologist and try again for a diagnosis. While waiting for this appointment we had to get Alaysha a walker.

I also started doing my own research on the different kinds of ataxia. I found ataxia telangiectasia and felt as if I were reading a horror story about my baby. And I knew this was what she had. I

told the neurologist what I thought; he agreed so we had the test done and sure enough she has A-T!

I was kind of prepared for this but it's still like a nightmare come true. As soon as we knew what she had we decided to start fundraising for Alaysha so that she can get to experience as much as possible in her short life.

Her older brother Colton did a fundraiser at his school and raised \$600 for the National Ataxia Foundation. He also got us on the news to bring awareness of ataxia to our city.

We started a GoFundMe.com personal fund-raising website and put donation jars around town. Unfortunately some of them were stolen last week, but karma will take care of that.

We got Alaysha her own post ►►

office box so people can send cards and things. She also has a Facebook page called “Hope for Alaysha” where people can talk to her. She posts lots of videos and pictures.

The only way I can deal with this horrible diagnosis is to only think of today. Alaysha’s tomorrow isn’t promised. She is the happiest child. She always was and still is. Alaysha is the most thoughtful child I’ve ever met; she always thinks about how things will make others feel. Her smile can light up a room. Everyone knows and loves Alaysha. I’ve always taught her that everyone is different and she takes it to heart so it doesn’t bother her that she is different. I’m so thankful for that.

Life is a lot different now. We used to have a very laid-back, easy-going life. Now life is very

busy, filled with doctor appointments, therapy, worries and fear. But my daily goal will never change: that is to make sure Alaysha goes to bed with a smile on her face. Life isn’t always fair to everyone but we take the hand we’re dealt and day by day do the best we can.

I would never be able to deal with all of this without the support of family and most of all my soul mate David. We were brought together for a reason and now I see why; he is a wonderful dad to Alaysha. Something she has never had. We have a few challenges right now as her A-T is getting worse. She will need a wheelchair soon so we desperately need to get her in a one story house. A-T is a horrible condition but we do our best to not let it rule our lives. ❖

How You Can Help

As you read the stories of these children and their parents who live with the challenges of ataxia, please consider making a donation to the National Ataxia Foundation.

Established in 1957, the NAF is dedicated to improving the lives of persons affected by ataxia and their families through

support, education, and research.

The impact that your donation has is profound and far reaching. Research gives hope to these families, that one day there may be treatments and a cure for their children.

Donations can be made through our website, www.ataxia.org. Thank you.



Angela's Story

Story provided by Angela's Mom and Dad

We have three children, all beautiful girls; Angela is our middle daughter. Our world was turned upside down when Angela was diagnosed with Spinocerebellar Ataxia type 17. I can still recall how the oxygen left my brain and the sick feeling in my stomach on that painful day. A wonderful nurse had taken Ang for a walk in the hospital as we sat numb in the doctor's office while being told the devastating news of our daughter's future. The doctor told us to be strong in front of our children and cry at night when they were in bed.

Angela was 15 years old when she was diagnosed with SCA 17. Today she is 23. She does not seem that old though. The progression has caused her to no longer be able to do most things. It's almost as if her life stopped in its tracks. Angela cannot walk anymore, she is

in a wheelchair that we push because she is not able to operate it. Harness straps are necessary at all times. Ang cannot support herself sitting or standing. A headrest must always be in place otherwise her head falls backwards and she is not able to lift it up on her own. Choking occurs quickly.

Vomiting had become a very serious problem for Angela. We tried for months to figure out why this was happening. We were scared. Losing Angela certainly crossed our minds during this difficult time as she dropped down to 69 pounds. We eliminated foods, added foods but nothing stopped the vomiting. We decided to take Angela off all medications, the weaning process took quite some time but eventually the vomiting completely stopped! She now also sleeps through the night or is at least ►►

peaceful. Unfortunately a large pressure wound near her tailbone opened up causing Angela great distress. The wound became infected. We added an air pressure mattress to her bed, a ROHO pad to her wheelchair, tissue nutrients to her diet and Angela was repositioned every 30 minutes. It took a little over a year for the pressure wound to completely heal. This was an incredibly painful time for Angela and also for her family.

It is necessary for us to feed Angela. Each meal takes one-and-a-half hours. The food needs to be chopped up very fine and have a high calorie count. Every morning her dad gives her yogurt, cereal with whole milk, Juven (which promotes tissue growth and strength), Karo syrup and of course ice cream. Every single morning without fail he does this. We need to be diligent in our efforts to feed her so she can at least maintain her low weight which is now in the 80's.

On a really good day Angela is able to say one word, it tends to be a one syllable word. This does not happen very often, to say once a month might be exaggerating.

Angela will try her best to nod for us. But there are times when she's just not able to do that and times when we are not able to tell if it's a "Yes" or a "No." Angela's losing her ability to talk has been one of the hardest things. Is she hungry? Is she in pain? Is she cold? Is she sad? Is she scared? We can only hope she knows we are trying our very best to give her what we think she needs and wants.

Angela is now very stable; she is tough, she's a fighter. Better yet, she is happy and likes to be near us. Her smile is as beautiful as ever! We continue to wait for a cure for Spinocerebellar Ataxia. We are so fortunate to have

“
**Angela is now very
stable; she is tough,
she's a fighter.**
”

so many loving and dedicated doctors, nurses, counselors, researchers and all of the others who tirelessly provide their time and talents to help make life better for others ... to find a cure. Please don't give up, please don't be discouraged. When you find that cure, our daughter will be able to again say the words that we so long to hear, "I love you."

Thank you from the bottom of our hearts,

– Angela's Mom and Dad ❖



Cameron's Story

Story provided by Cameron's Mom

Cameron was diagnosed with ataxia at the age of six. He is nine years old now and still being tested. At the time he was diagnosed, I chose to not find out the type of ataxia that he has until recently when I contacted a geneticist. Cameron first started showing symptoms at age 11 months when he started walking. He would fall more than he should have. Everyone told me, "Oh it's normal," but as his mother I knew something was wrong.

At the age of three he went to "Early On" for physical therapy, occupational therapy and speech therapy. Cameron has been through so many specialists and appointments, that I have thought that is all that life consists of. I have two other children who are completely fine and do not show

any signs of ataxia. We try to make life as normal for them as possible. They have both been brought to so many appointments with us, especially his brother Caiden. He puts meaning to the phrase, "My Brother's Keeper." He is always tying Cameron's shoes, helping him stay steady while he dresses and buttoning his clothes.

As a mother you don't want to see your child struggle to do normal everyday things like walk, talk, button, ride a bike, and eat. Cameron loves to sing and dance and wishes to play sports, but unfortunately ataxia has taken that from him. I have many days where I want to cry and feel angry, but Cameron keeps me going, because he is a bright, funny, beautiful child with a heart of gold. ❖

Visit www.ataxia.org for more information



Liesel's Story

Story provided by her Mom and Dad

Liesel was diagnosed in June 2014 through genetic testing as having Spinocerebellar Ataxia type 29 (SCA 29), an extremely rare disease. Her gene mutation is the first doctors have seen.

We started to notice something was different when she was around seven months old. She was delayed with all milestones: not sitting up until nine months, not crawling until 14 months, and a very unstable walk followed at 26 months. She began physical therapy and occupational therapy at 10 months and at 15 months began speech therapy. Currently she receives six hours of therapy a week. Our pediatrician referred us to a neurologist at 15 months old. The journey took over a year-and-a-half before all the genetic testing came back.

After countless hours of research and visiting doctors, Liesel is

currently under the watchful eyes and care of neurologists and neurogenetic doctors at UCLA Children's Hospital. We will be seeing doctors with the ataxia program at Johns Hopkins Hospital in July.

Liesel loves to play with her older sister Reese, who is six years old. She giggles with pure joy when entertaining others with her singing, or in her dance and gymnastics classes. She is social and loves to be around people, attends school for several hours each day. Her smile will certainly brighten your day.

Liesel faces daily challenges; her speech is delayed of that of a two year old, making it extremely hard to understand her needs and wants. Recently we saw a gastroenterologist due to swallowing

Continued on page 8

concerns. We have found that she coughs when drinking water and she is at high risk for choking. Walking is still so hard, her gait is very wide causing her to be unbalanced and she falls often. Both of her legs are very weak. Most of the time we carry her places or she rides in a stroller.

In the last year Liesel has begun to have seizures, although we suspected this months before her first seizure. We have found these seizures, as they become more frequent and intensify, have affected her walk and speech. For

days and weeks after seizures she is at a greater fall risk or may not walk at all. She tires easier and is in need of a walker to travel the halls of school. Liesel's doctors have ordered her a walker to use at home as well.

As her parents, we hope for a cure. It is difficult to watch her struggle; we don't know what to expect in the future but continue searching for doctors and researching this disease. We feel so alone, often finding ourselves informing doctors and specialist across the USA about SCA 29.

We pray that we are making the right decisions with her care. ❖



Maya's Story

Story provided by her Dad

About a year-and-a-half ago we realized that (our now 11-year-old daughter) Maya's "differences" were not normal. When I held Maya's and her brother's hands on the way to

class, it seemed weird that Maya always seemed to tug and pull on my hand. It was such a contrast to the way her brother held my hand. My mom commented that Maya seemed "off balance" and ►►

we should get her checked by a neurologist.

After months, we finally had an appointment. The neurologist did a couple of basic tests as part of his examination (finger to nose, hop on one foot, walk across the room heel to toe) and it was clear that something was very wrong.

The neurologist thought that the bouncing of Maya's eyes was indicative of ataxia telangiectasia, however, the blood work said no. The Athena Panel for known forms of genetic ataxia eventually came back inconclusive. After a year of tests and a trip to Massachusetts General we finally had a diagnosis: Atypical TPP1 Deficiency, or SCAR 7.

Maya's condition is caused by mutations in the same gene that causes late-infantile neuronal ceroid lipofuscinosis (a form of Batten disease). Those of you in the ataxia community will recognize SCAR 7 as Autosomal Recessive Spinocerebellar Ataxia Type 7, a type of ataxia reported as occurring in six of 12 siblings from a Dutch family and one

other unrelated Dutch woman. Guess what ... Maya is not Dutch, yet she is the eighth reported SCAR7 case in the world.

She is said to have a mild phenotype, however, there is nothing "mild" about watching your child slowly lose her ability to walk ... to watch your child struggle with her "differences" in school. "Mild" provides little comfort, when she falls, or when there is a

new scar or when she cries, because she doesn't understand why her body is revolting against her.

In order to find a cure/help for Maya, we need to find more kids and adults with Maya's exact

phenotype/diagnosis. The more we find, the more science, medicine and the respective communities will pay attention and join the fight. I suspect that there are a number of children and adults with Maya's symptoms who are simply undiagnosed but have TPP1 mutations. If you are out there, then we need to find you. Please contact me at bjames358@gmail.com.

“
**In order to find a
cure/help for Maya,
we need to find more
kids and adults
with Maya's exact
phenotype/diagnosis.**
”



Olivia's Story

Story provided by Olivia's Mom

Our daughter and first child, Olivia Virginia, was born without complications in 1994. However, by the time she was supposed to toddle, her father and I realized that there was something wrong. She could not stand or walk independently. She drooled abnormally at times, her speech was slow, and she was shaky. Her pediatrician referred us to a neurologist who looked perplexed and said that she had some form of cerebral palsy.

I was devastated realizing that doors would be closed to her. Over time, we would come to appreciate all that Olivia could do instead of focusing solely on her limitations.

At one point early in Olivia's treatment, we were advised to see a pediatric physiatrist who specializes in Physical Medicine and Rehabilitation. The doctor was a

great asset in our quest to help Olivia. When she came into the room to see Olivia for the first time, she was blowing soap bubbles and immediately won the heart of our little girl. Through the doctor, Olivia was fitted with a K-Walker, a mobility aid. For the first time, Olivia was able to walk without someone helping her. It was a tearful moment for her parents but they were tears of joy.

A neurologist at Children's Hospital in Columbus told us that Olivia had ataxia. Through the National Ataxia Foundation's web site, we found a doctor who had an interest and background in ataxia. We drove from Ohio to Chicago to meet the doctor and gain his opinion of Olivia. He said that she would probably parallel normal but continue to have a disability. ►►

Olivia underwent physical therapy to help her with movement, and had several MRIs as a young child. It was discovered that Olivia's cerebellar vermis is abnormally small. Damage to the vermis can cause cerebellar ataxia.

Olivia was diagnosed with spinocerebellar ataxia. Later, however, genetic tests were negative for a link to Olivia's ataxia. It is quite possible that Olivia's ataxia was caused by an environmental factor.

As parents, we were becoming familiar with the world of disabled children, particularly when Olivia became a member of the ballet company, "Firebird," in Dayton, Ohio. They sponsored a ballet with prima ballerinas and children who used wheelchairs or walkers. Olivia was one of two young girls wheeling their walkers on stage. The ballet was fabulous for the children who could "dance" behind the stage lights and hear the thunderous applause. The ballet involved the extremes of physical ability and inability, but fostered joy and mutual

appreciation as the two came together in a classic ballet.

When the National Ataxia Foundation held its annual conference in Tampa, Florida, Olivia and I attended. I had more questions about ataxia than answers, so this was a fantastic opportunity to hear from experts in the field. Olivia had congenital (since birth), sporadic (unknown cause), ataxia and I asked the panel about

her condition. While Olivia sat on the floor coloring, I learned that there was not a cure for her disease and my heart sank. However, I was not going to let my disappointment and sadness affect my little

girl, who was generally a happy and active person. My husband's positive attitude helped me cope as well.

Through the National Ataxia Foundation, we found a wealth of information and strength in a local support group. For parents learning that your child has ataxia, I would highly advise you to attend

“
**When the National
Ataxia Foundation
held its annual
conference in Tampa,
Florida, Olivia
and I attended.**
”

Continued on page 12

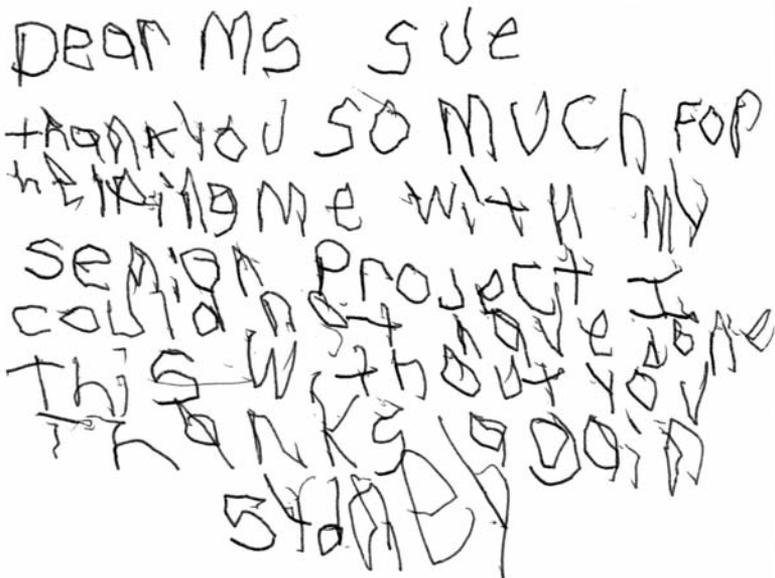
Continued from page 11

a national conference or join a support group. Never give up hope that your child can accomplish more than they are told they can. At times Olivia was left behind by her counterparts or her young, lively cousins, but her spirit is strong and she has stayed positive throughout her life. A strong spiritual foundation has kept her grounded and has led to friends and mentors.

Olivia graduated with a regular diploma in 2014 and it was a super day for her and her family. And how is she now with ataxia? She speaks and processes information slower than normal, and she falls occasionally, but she keeps improving as she continues to understand how to move. She enjoys art and will study graphic design. Despite her ataxia, Olivia is on the road to success with her will to succeed and the many support systems available. ❖

Handwritten note by a teenager with ataxia

Ataxia often causes motor skills to deteriorate. Here is a note written by a teenager with ataxia. The note reads: “Dear Ms. Sue, Thank you so much for helping me with my senior project. I couldn’t have done this without you. Thanks again. Sydney”



DEAR MS SUE
THANK YOU SO MUCH FOR
HELPING ME WITH MY
SENIOR PROJECT I
COULDN'T HAVE DONE
THIS WITHOUT YOU
THANKS AGAIN
SYDNEY



Yasin's Story

Story provided by Yasin's Mom, Sakina

I'd like to introduce you to my 11-year-old son Yasin. When he was six years old, he was diagnosed with Friedreich's ataxia (FA). Having FA causes challenges for Yasin: difficulty walking, getting dressed, writing, eating, sitting and standing. To assist him with sitting and standing he uses a back brace and leg supports, as well as ankle-foot orthosis (AFO). For mobility, Yasin requires a walker or wheelchair when he is more fatigued. He needs help doing most physical activities.

Like most kids his age, Yasin's favorite activities are reading and playing video games. Yasin also participates in a sports program for individuals with special needs. A few years ago Yasin advanced to basketball, playing with a special wheelchair that enables him to move around the court fast.

As a parent of a child with

ataxia, I face many challenges. One of the biggest challenges with FA is that Yasin's needs are constantly changing. The most recent struggle has been to ensure a safe surrounding by removing obstacles and purchasing various medical equipment to help reduce harm to our bodies, both my child's and mine (when assisting him). Another challenge I face is advocating for his best interest. There are so many complexities of FA that many people don't understand. I feel that the more that is known about the disease, the better people can be in helping.

I work hard to ensure that he doesn't lose his childhood to a disease that is already taking away from him day by day. I try to ensure that he feels included, happy and loved. I am always praying for a cure and hoping that we put an end to Friedreich's ataxia! ❖

In Memory...

Alan's Story

Alan was diagnosed with Spinocerebellar ataxia type 7 (SCA 7) in 2005 when he was 7 years old. His first neurologist identified that Alan had ataxia – and genetic testing results confirmed SCA 7.

Alan continued his care at University of Texas Medical Branch (UTMB). His neurologists there were ataxia specialists which was a huge blessing to me because they knew a tremendous amount



Alan

about SCA 7 and were able to share that information with me. This allowed me to respond to Alan's medical situation appropriately.

Alan enjoyed any sport that involved running. He enjoyed riding his bike, swimming, climbing trees, playing basketball, and soccer. Alan was able to do all these activities until he turned 7. His vision declined from 20/20 to going blind when he was 11. At the same time Alan's mobility was declining. He could no longer

run, climb trees, or swim. At age 8 he was using a walker and by the age of 10 he was using a wheelchair. Even to Alan's final days he continued to ask me to help him to walk.

The most difficult challenge I faced as a parent with a child with SCA 7 was that there was nothing I could do to stop the progression of the disease or get rid of SCA 7. If we had not had access to excellent medical and support services I would not have been able to be the best mother I could be for Alan. His medical support consisted of his pediatrician, the ataxia specialist physicians, a gastroenterologist, a physical medicine doctor, an ophthalmologist, a cardio-respiratory doctor, and an excellent whole health nutritionist. I also utilized services from DARS (Department of Assistive and Rehabilitative Services) and a support group for parents with children with disabilities.

I would recommend doctors share the following with parents or guardians of children with SCA 7:

- Due to the progressive nature of SCA 7, adjustments to your ►►

child's physical condition are on-going. Also suggest that they work with an ophthalmologist who specializes in low vision and retinal muscular degeneration because with SCA 7 the patient eventually goes blind.

- Because muscle coordination is decreasing, adjustments need to be made regarding how the individual with SCA 7 meets their dietary needs. Alan was able to

masticate his food but soon needed a feeding tube as his main source of caloric intake.

- Stress the importance that time is of the essence. If there are experiences and activities the child wants to do, try them as soon as possible due to the degenerative nature of the disease.

- Finally, recommend that they utilize NAF.

– Alan's mother ❖

NAF Is Here to Help

The stories you have just read about children who are affected by ataxia were generously shared by their parents and loved ones to better equip clinicians as they provide medical services to their patients and to give a human face to the disease that ataxia researchers study in their labs.

The National Ataxia Foundation (NAF) is a non-profit membership organization that strives to improve the lives of those affected by ataxia through support, education, and research.

Support – Local ataxia support groups are facilitated by NAF so that families can connect with one another. In addition, the Foundation offers an annual membership meeting to provide comprehensive information about ataxia by experts in the field of ataxia research, genetics, and

medical care.

Education – NAF has developed an extensive library of ataxia related fact sheets, books, and videos. Also available to its members is *Generations*, the Foundation's quarterly news publication. NAF also offers a resource list of neurologists who specialize in ataxia and other movement disorders.

Research – Through NAF's research program, the organization provides funding for promising ataxia research studies. NAF also supports ataxia research by promoting participation in patient registries, clinical drug trials, and natural history studies so that viable treatments can one day be a reality for those affected by ataxia.

For more information about NAF visit www.ataxia.org.

About NAF

The National Ataxia Foundation (NAF) was founded in 1957 as a non-profit, charitable organization with a mission to find the cause and cure for all types of ataxia.

The focus and purpose of the Foundation is to support promising research and provide meaningful programs and services to those affected by both dominantly and recessively inherited ataxia as well as sporadic ataxia and other closely related conditions.

The Foundation's objectives include:

Empowering ataxia families and persons at risk

NAF encourages a complete neurological examination to provide an early and correct diagnosis. NAF maintains a current referral list of Ataxia and Movement Disorder Clinics and neurologists who are familiar with ataxia. NAF provides assistance to start and sustain support groups and provides referrals to groups where available.

Increased awareness and education about ataxia

NAF assures that accurate infor-

mation will be available through a variety of educational programs for those affected by ataxia, physicians, genetic counselors, physical therapists, other health professionals and the public. Through literature, an in-depth quarterly news publication, and a comprehensive website, NAF creates awareness and serves as a resource for current ataxia information.

Prevention of ataxia

At this time, there is no treatment available that can prevent ataxia from developing in a person who has an affected gene. NAF encourages genetic counseling to help families make informed decisions about family planning and genetic testing.

Ataxia research activity

NAF continues to promote and fund world-wide ataxia research designed to better understand all types of ataxia, to find the genes that cause ataxia, and translate this information into treatment methods.

More information

More information can be found on the National Ataxia Foundation's website www.ataxia.org. ❖

National Ataxia Foundation

2600 Fernbrook Lane, Suite 119
Minneapolis, MN 55447-4752

Phone: (763) 553-0020 • FAX: (763) 553-0167
Internet: www.ataxia.org • E-mail: naf@ataxia.org

©2015 National Ataxia Foundation. All rights reserved.