# Children with Ataxia


NAF
National Ataxia Foundation
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# Children with Ataxia

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## 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.

# Izzy's Story

*Story provided by Izzy's mother*

Isabel (Izzy) was diagnosed with Friedreich's ataxia (FA) in July 2010, at the age of 9. Her symptoms however began at age 5, and we had many years and many doctors' appointments before all the pieces of the puzzle were finally put together.

Izzy has had many favorite activities, they change as she changes. At age 6, she loved to hula hoop, and once won a hula hooping contest! At age 7 she fell in love with art and drawing. While in second grade she joined a baton twirling group and marched in our local parade.

As her FA has progressed these things have all become increasingly more difficult for her. The hula hoop has been retired, this year she was in the parade with her baton group, but sat in the truck and waved and smiled at all the spectators. She is unable to walk unaided now, and requires a walker or electric scooter. She doesn't draw very much anymore, her motor skills have deteriorated significantly and she gets frustrated and sad when her pictures don't come out the way she envisioned.

At school during recess, she used to love to jump rope with her friends. Now she spends her recesses in the health clerk's office laying down for a rest. Just so she can get through the rest of the day.

As a parent of a child with ataxia, you live in a constant state of heartbreak. You push it aside and put on your happy face and get on with life, and you make each day count. But it's always there in the back of your mind. We still talk about what she will be when she grows up, and I tell her she can be anything she ▸▸

wants, it will just be a much harder journey for her. I've held her many, many times while she cries and tells me it's not fair. It's not fair. But it is what we've been handed and we will make the best of it. One of our biggest challenges as a family is with our "normal" child. He is four years younger than Izzy and can ride a two-wheeled bike, run, and play baseball – all the things that Izzy wishes she could do, but no longer can. He loves her and helps her out all the time. It makes me sad that he worries about her and that his childhood is complicated by a sibling that requires a lot of attention. He has been dragged to a hundred therapy visits and doctors' appointments. But more than anything to know that he will face losing his big sister someday is unbearable to me.

It took us four years to get her diagnosis. We live in a part of the country where FA is less common. Our neurologist polled all his colleagues and between the five of them they'd only had one other case. Those four years were extremely frustrating, and put a large financial burden on our family. We paid out of pocket for neuropsychological exams, physical therapy and occupational therapy. We knew that something wasn't quite right, but we kept being reassured that she would be fine. Since she didn't have a definitive diagnosis our insurance didn't pay for quite a bit. In the year since her diagnosis she has had to pick out a walker, and an electric scooter. These are not things that 10-year-old little girls should have to worry about picking out. But she has and they have made her life so much easier.

> **"As a parent of a child with ataxia, you live in a constant state of heartbreak. You push it aside and put on your happy face and get on with life, and you make each day count. But it's always there in the back of your mind."**

We have our fingers crossed and keep hope alive that someday a treatment or cure for ataxia will be found. ❖

# Angela's Story

*Story provided by Angela's parents*

Our daughter's name is Angela. She was diagnosed with Spinocerebellar ataxia type 17 (SCA 17) when she was 15 years old; Angela is now 19 years old. The past four years have been filled with sadness, helplessness and frustration. We are fortunate; our family has not been torn apart by this devastating disease. It has perhaps made us stronger in some areas.

Angela does not care to attempt anything too physical. Everything is so hard for her and she has a great fear of falling and getting hurt. She is most content watching movies or television shows. Anything from "Animal Planet" to cooking shows, cartoons to superheroes and comedies to love stories. When she is watching she becomes so focused on the show that she can block out almost all of her surroundings. Another pastime she loves is using her iPad. Angela enjoys the games very much and she is able to use the touch screen very well.

We no longer ever leave Angela alone. Last year she could still walk with a walker, but now she can only walk with our personal assistance (20 feet on a really good day). If Angela sits up in bed during the night she can no longer lay herself back down. Telephones can no longer be used. We can still understand her speech but some sentences require numerous repetitions. Her writing is no longer legible. Angela has difficulty sleeping at night and is never able to nap during the day which makes her even weaker. The aid of a mild sleeping pill in the evening has helped with this problem. There is shaking in her hands and feet, but these symptoms are not always ▸▸

present. Most of the dystonia is under control by medication.

Angela does physical therapy at home two or three times a week. Some sessions go fairly well and others do not. If she is the least bit tired or under the weather everything is such a struggle for her. The simplest things in the world to you and I are almost impossible for her. Angela has incontinence problems, frequent urges to go to the bathroom along with accidents. Medication seems to help on some days but on other days it's as if it's not working at all.

Angela has two healthy sisters. One is younger and the other older. Our 22-year-old daughter sometimes worries about the hereditary factor of SCA 17. Being tested scares her even more. We know of no other family history of SCA's. We work very hard at making the lives of our two healthy children "normal." But there's no doubt it has affected them. As parents we do not want to give up. Sometimes we feel alone in this fight.

Last year we put a pool in with the hopes of some great times for Angela and her sisters, along with some good physical therapy. But the progression from last year to this summer has not gone unnoticed. It certainly has made it harder and Angela gets cold so quickly. One pool day may require two recovery days.

> **"The past four years have been filled with sadness, helplessness and frustration. We are fortunate; our family has not been torn apart by this devastating disease."**

Angela can feed herself but tires after 30 minutes or so and even then her plate is barely one-quarter empty. She'll say she is full but really she's too exhausted to finish. So then we take over spoon feeding her. She needs the food for energy and also the calories; she weighs only 84 pounds now.

Angela's dad walks her every day in hopes of her maintaining her muscle mass. We want her to be ready when there's a treatment or better yet a cure. We know if we do not make these efforts Angela will spiral downwards rapidly. So we have decided to be as strong as we can for her. ❖

**Roman pictured dressed for a "fight ataxia" awareness and fundraising event**

# Roman's Story

*Story provided by Roman's grandmother*

I am the grandmother of Roman, age 12. Roman was thought to be a very normal baby and toddler. Our first hint that something was wrong was when he played baseball, around the age of 5. He would slide into every base because, as we know now, it was easier for him than running. He would get so tired that the coach would jokingly carry him into the dugout; he was the smallest on the team. The family just thought it was because he was so small that he had a hard time keeping up.

It took three years to diagnose Roman with Friedreich's ataxia (FA). After his doctors discovered he didn't have knee reflexes they went through a process of elimination to determine the cause of his problems. The final diagnosis came in 2008, when Roman was 8 years old.

As I am sure you can imagine it was a devastating diagnosis and something that the family is still coming to terms with. It all seems so surreal.

Roman is filled with hope that there will be a cure and that he will be someday be "normal." He uses a walker and as you must be aware, middle school students are less than understanding and compassionate and he gets his share of being made fun of. This is tough to hear and I hurt for him but he just gets tougher and smiles even more.

Roman's parents have done all that is necessary to make life a bit easier for him and as his greatest advocate, I have gotten on the parents FAPG (Friedreich's Ataxia Parents Group) email system to hear how others cope so I can suggest things to Roman's parents.

We have had two fundraisers ▸▸

at Outback restaurant and Roman was "King for a Day" as we raised money for research. He will be attending MDA camp this year and is looking forward to it. We have also participated in two annual MDA walks. We are always the biggest group, topping 65 people last year, and again raising money for research. The fundraiser's raise Roman's awareness of just how many people love him and want to see a cure for FA.

Roman is a trooper. He lives one day at a time, as do we, his family. He is getting worse. He falls so much, and more often than not, hurts himself. I cry for him, but I have praise for him as he endures this devastating disease. He is encouraged that there will be a cure before he has to use a wheelchair. He is willing to participate in any trial that comes about for FA research. He is willing to be a guinea pig for the cause, so he says.

Roman is a great video game player and has many Xbox friends. This is his passion and his parents encourage him to start thinking of ideas for games so he can go to work for Microsoft or another company. As much as he loves his Xbox, he misses baseball and outdoor sports and still tries to join his cousins when they play. He does pretty darn good and they all applaud him as they help him around the bases.

One day I picked up Roman from school and a school worker told me that Roman stood up in his health class and explained to the students that he has FA, a disease that makes him walk like he is drunk and that there is no cure – yet. He did this on his own and I have never been so proud of him.

> **"Roman is filled with hope that there will be a cure and that he will be someday be 'normal.'"**

There is a reason for everything that happens in our lives and Roman will come through this. With the help of research and dedicated doctors I know a cure will be found before it is too late for Roman. Thank you all for your devotion to finding a cure for FA. Please hurry up! Roman wants to play baseball and run around the bases, without help. ❖

# Jordan's Story

*Story provided by Jordan's mother*

Jordan was diagnosed with Spinocerebellar ataxia type 2 (SCA 2) when he was 7 years old. He was in the first grade and struggling to learn how to read when his teacher noticed his eyes didn't want to move with the words on the page.

Up until that point Jordan was like any other first grader. He rode a two-wheeled bike, ran, jumped, climbed and played baseball. He had a great sense of humor and loved to laugh and make everyone around him laugh.

Over the last six years we have been watching the disease take all that away from Jordan. The hardest thing for him was four years ago when he had to get a walker. He would rather fall than have to use the walker, but then the falls became more frequent and more painful. He now has scars all over his body from the numerous falls, and twice he chipped out his front teeth. Reluctantly, he started using a walker. Then two years later he began getting fatigued from having to use his walker and we had to get him a wheelchair.

From September to May he lost over 20 pounds. He would try to eat but many times he would aspirate on the first bite and not want to eat anymore. In May 2011 Jordan had his first surgery, which was having a G-tube placed.

Everything was going well for six weeks; he was gaining weight and even had more energy. But by the middle of July everything changed when he started having severe abdominal pain. He was admitted to the hospital where they did numerous tests. The results were normal and there were no answers for his pain. The G-tube was removed and a GJ-tube was placed instead. ▸▸

The challenges Jordan faces are constantly changing. The things he can do one day, he may not be able to do the next. Today he struggled to stand without having his knees buckle, struggled to breathe without gagging on phlegm, struggled to drink juice without coughing, and struggled to make it through the day without getting bruises from his limbs jumping all over uncontrollably. Now, Jordan is unable to talk so communication is very difficult.

As a parent, I am faced with many challenges that most parents don't even think about. I need to figure out which things he won't be able to do each day. Also, my husband has SCA 2, and due to his condition, he is not able to help out in the care for Jordan.

Most medical equipment is not made for Jordan, so we need to improvise almost everything. I am constantly fighting the insurance companies to get supplies paid for or to get them in a timely manner. We had to wait three months for them to approve a wheelchair!

Five years ago I asked our neurologist how much longer Jordan has. He said three to five years, and he made it very clear that we should not dwell on the end, but live every day like it was our last. That is what we have been doing. Every day we try to do something fun; something that Jordan will enjoy. We don't know what to expect from one day to another or if we even have another day. There is nothing we can do to stop this disease. There are no cures or treatments. Even the knowledge about the disease is very limited, especially in children.

> **"Every day we try to do something fun ... We don't know what to expect from one day to another or if we even have another day."**

Jordan's mind is still good and he understands everything we say but his body is giving out. Most of the things he enjoyed doing a year ago he is unable to do now. Now he enjoys watching TV, listening to music, having books read to him, riding on an ATV, hanging out with family and friends and playing pranks.

Ataxia has taken a lot away from Jordan but he still has a smile for everyone. ❖


# James' Story

*Story provided by James' mother*

James is a strong-willed 5-year-old boy who has Undetermined Spinocerebellar ataxia. At the age of 10 months I noticed that James was showing lack of balance when trying to sit independently, this is when the journey began. Appointments started at the Development Delay Clinic at Children's Hospital. We were then referred to Pediatric Neurology who also noticed some type of delay with James, one which they were not able to put their finger on. At this point I had started James in weekly therapies (physical, occupational, and speech).

The MRI showed severe atrophy in the cerebellum. I was advised to come to the office where I was told that James has Spinocerebellar ataxia. I was told that there are several types – all with different outcomes. I was told James would never be able to walk or stand on his own, and may have issues with smiling, swallowing, blinking, etc. My heart hit the floor.

Thereafter, I made the decision to go to the Cleveland Clinic to get another set of eyes on him. The neurologist reviewed James' MRI and stated there was more testing we could do – but it seemed as if James was slowly getting better – through therapy. We have completed the ataxia panel testing (James did not fit into any of the groups). Several tests have been run through the Genetics Department at the Cleveland Clinic. None of which has been able to tell us the cause or type of ataxia that James has.

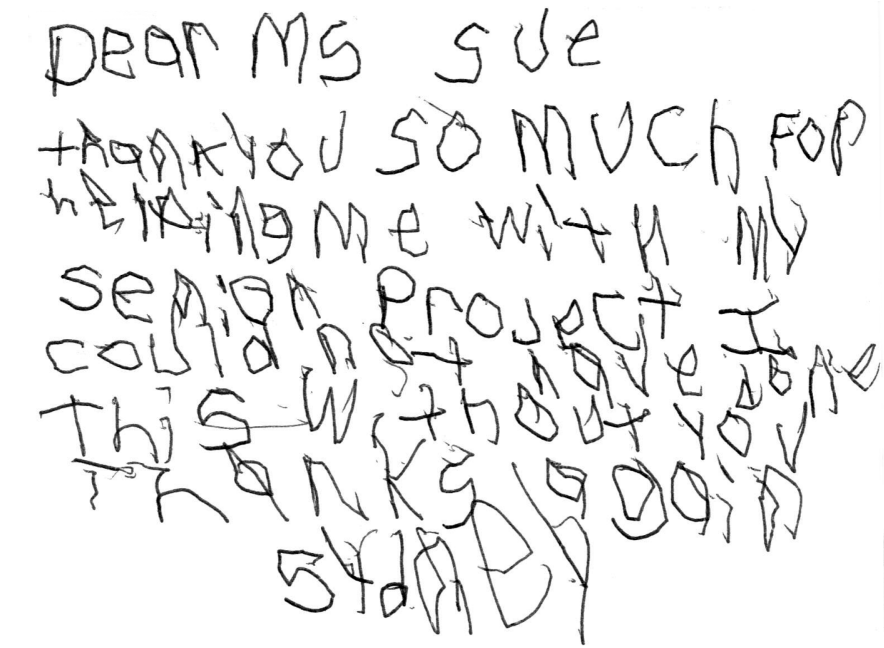
As of today James is able to walk using a walker and by holding onto the countertops and the back of the couch, or while holding ▸▸

on to someone's hands. He wears AFO leg braces to help support his ankles – he is also able to use his wheelchair. The wheelchair is mainly used for long walking trips, shopping sprees, and bus rides to and from school. James has started kindergarten this year and has been mainstreamed into public schools. It seems that James' only delay is with the physical movement.

I hope and pray he keeps moving up the ladder and not down. I hope he is able to walk on his own one day. Regardless of knowing what the prognosis is or not – I am taking this one day at a time. I have never seen a child with as much strength and determination as he has. It is amazing to be given such a challenge in life at such a young age and to be one of the happiest little boys I have seen. ❖

## 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"*



# In Memory...

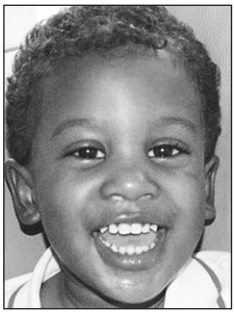
*The following stories are about children who lost their struggle with ataxia. The children's parents recount their journey with ataxia – the devastation, the glimmers of hope, and the important role of the medical team in caring for their child.*

## Jordan and Sydney's Story

Brother and sister Jordan and Sydney were both diagnosed with Spinocerebellar ataxia type 7 (SCA 7). The children's mother shared the following about her family's struggles with SCA 7.

### Jordan's story

Jordan was diagnosed with SCA 7 at age 2, and passed away from its complications at age 3. While he was with us, Jordan enjoyed playing with a ball and listening to music.



**Jordan**

As his illness progressed, Jordan had problems eating, walking, and seeing. Daily activities were a challenge – feeding required a feeding tube and bathing required a bathing chair. It was also difficult to try to find ways to keep Jordan entertained as his mind was still intact; he was just unable to do anything.

As a parent I struggled to provide the best care for Jordan while adjusting to his rapidly changing symptoms and abilities. Every two months we were dealing with constant change – it was like trying to hit a moving target.

### Sydney's story

Sydney was a very active and outgoing child. She loved singing, making s'mores, and playing Uno cards. Sydney was diagnosed with SCA 7 when she was 4 1/2 years old and passed away at age 8. We realized something was not right after she experienced frequent falls, choking when eating, and was unable to see and speak. She became very frustrated, but never gave up.

**Sydney**

As a parent of a child with ▸▸

ataxia I struggled with questions like "Mommy, will I ever walk again?" which was a question Sydney asked a few months after using a gait trainer. Soon she was unable to walk at all. It was also a struggle to know that this disease would prematurely end her life, as it did her brother Jordan's.

**Message from a Parent**

As a parent of two children with SCA 7 the most difficult physical problems we faced were feeding, communicating, walking, and vision.

*Feeding* – Feeding tubes did not work too well because it was difficult to find the right formula that the kids could keep down. I think we went through every formula on the market without success. I eventually found something online called "Reliv Now for Kids," a nutritional supplement that I mixed with goat milk, which worked really well. Sydney had to get a GJ-tube, bypassing the stomach completely because she couldn't keep anything down and wretched uncontrollably.

*Communication* – I'm not sure why both kids became nonverbal; they just made sounds – different moans, unable to make any words. I asked doctors and speech therapists, but they were not able to give a definitive answer. We used a communication device with one of the kids called "Go Talk 2," where you record common phrases the child uses and each phrase has a picture to represent it. This helped for only a few months because as the ataxia got worse, the spasticity in the hands made it too difficult to push the buttons.

*Walking* – We started out holding hands for assistance, and then used a gait trainer, and eventually a wheelchair. Solutions were very short lived as there was constant change.

*Vision* – Ophthalmologists and low vision eye doctors eventually could not support the various vision problems the children had. We were referred to a neuro-ophthalmologist which involved running lots of tests but we were unable to do anything about what was occurring. It was not really helpful. We went every six months for a check-up until their vision was completely gone.

This is a horrible disease. It is frustrating to see your child is dying and no one in the medical community knows what to do other than treat the symptoms and monitor the progression. I pray that someone will find a cure soon.

*– Jordan and Sydney's mother* ❖

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

**Alan**

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 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 information 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**
600 Highway 169 S., Suite 1725
Minneapolis, MN 55426
Phone: (763) 553-0020 • FAX: (763) 553-0167
www.ataxia.org • naf@ataxia.org