The Voices of SMA

The Experiences of Individuals and Families and their Views on Living with Spinal Muscular Atrophy
Messages from SMA Families to the FDA

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“A key part of regulatory decision making is establishing the context in which the particular decision is made. For purposes of drug marketing approval, this includes an understanding of the severity of the treated condition and the adequacy of the available therapies. Patients who live with a disease have a direct stake in the outcome of FDA’s decisions and are in a unique position to contribute to the Agency.”
– Food and Drug Administration (in announcing the Patient-Focused Drug Development program)

FDA has committed to obtaining the patient perspective. To assist the Agency in this effort, in April 2015, Cure SMA coordinated a meeting between key FDA officials and patients and caregivers of spinal muscular atrophy to:

• Expand the FDA’s understanding on priorities of our patient community in drug development and voice patient views.
• Describe the impact SMA on patients and their families’ daily lives.
• Describe meaningful change in SMA patients’ everyday lives.
• Describe the patient community’s preferences and expectations for an approved treatment.

As part of SMA Awareness Month in August, Cure SMA sought to further the dialogue between the SMA patient community and FDA through “Share Your Story,” an open-ended survey that allowed parents and patients to speak frankly to the FDA.
Objectives and Methods

The objective of the survey is to share stories from the community with the FDA and other stakeholders to help them better understand the perspectives of SMA families. Using an online, open-ended survey implemented on the Cure SMA website, we asked families, “If you had a chance to talk to the FDA, what you would want them to know?” Cure SMA reviewed each story individually and identified key themes from the entire set of stories and chose representative quotations. Concordance was assessed between stories and the results of a prior qualitative study to understand the views individuals with SMA, parents of those affected, and clinicians who specialize in treating SMA.

Results

More than 75 SMA patients and their caregivers participated in the “Share Your Story” survey. What follows is a selection of excerpts from these stories; an appendix containing the full set of stories can be found on the Cure SMA website.

Three key themes emerged from the responses collected from this project:

THEME 1

The Psychosocial Impact of Living with SMA

The psychosocial effects of coping with SMA are substantial and wide-ranging both for the individuals living with SMA and their family. Ten areas characterized the impact:

1) confronting premature death
2) making difficult treatment choices
3) fearing the loss of functional ability
4) coming to terms with lost expectations
5) fatigue and stress
6) stigma
7) limitations on social activities
8) independence
9) uncertainty and helplessness
10) family finances

Representations on the burden of SMA can be found in the stories provided in this report.
Defining Meaningful Change

The significance of any particular change was relative to where an individual was on the functional spectrum. Avoiding functional decline was voiced as having great importance. Small losses in functional ability could translate to devastating effects on the family. Even very small improvements in functional ability are viewed by the family as having enormous benefit, including critical implications for quality of life and independence.

- Meaningful change was relative to current abilities.
- Avoiding a decline in function is an important outcome.
- Small changes make a huge difference.

Other Critical Parameters for Measuring Change

Participants stated that items on the motor scales typically used in clinical trials accurately reflect features of SMA, but also noted that other aspects of SMA, such as strength, fatigue/endurance, respiratory function, and ability to perform activities of daily living should also be assessed.

- Participants view the Expanded Hammersmith Functional Motor Scale (HFMSE) positively.
- However, there is concern that the HFMSE may not be sensitive enough to detect change.
- Participants identified important abilities that were not captured by the HFMSE, including:
  » Strength
  » Endurance
  » Respiratory function
  » Independent movement
  » Activities of Daily Living
  » Communication
  » Quality of life
Our Stories

For each key theme, representative passages from stories were selected to provide context to the views of SMA patients and caregivers.

The Psychosocial Impact of Living with SMA

MY SON GAVIN WALKER KRESS WAS BORN ON MAY 1ST, 2006 AND DIED ON OCTOBER 3RD, 2006. When he was diagnosed with SMA Type I, I had no idea what the disease was, but I learned. I learned, and I reached out and I found an amazing network of families who were all fighting a similar battle.

I learned – just like everyone caring for an SMA patient knows – that conditions can change in the blink of an eye. One day, Gavin and I met friends to shop for an upcoming Halloween party, and the next he had stopped breathing and was rushed to the ER. He was in the PICU for 10 days before we lost the fight. **Gavin’s fight was short**, but I swore to him that I would keep fighting back against Spinal Muscular Atrophy, and nine years later, I am still wielding every weapon I have against SMA. Nine years later, and I have celebrated major milestones in the war, and mourned the loss of young fighters.

Nine years later, and SMA has affected my life threefold. In 2013, my cousin and his wife lost their second son – Ryker Don Conrad – to SMA Type I. In 2014, a friend’s son was diagnosed with SMA Type I and continues to fight. **Nine years later, and I have had to hold my daughter as she struggled to absorb that she has a brother that she can never meet. Nine years later, and I’m not ready to give up.**

> “...every second that a family goes without treatment for their child might be the last second that they see that child healthy.”

And so, FDA, I ask you to look at all of the progress that has been made in nine years, and I ask you to remember that every second that a family goes without treatment for their child might be the last second that they see that child healthy.

— Renee Hunter, mom of Gavin (deceased), SMA type I
Our daughter was diagnosed with SMA almost 13½ years ago. The process we underwent to find out what was affecting our daughter Jadelyn was painful & stressful not only for us, but for her. When she was diagnosed at 18 months old we did not know or understood what SMA was... at the time there was minimum information out there. There are several types & Jadelyn was diagnosed with type II, robbing her the ability to sit on her own, crawl, stand & walk. She has been confined to a wheelchair since she was 2 ½ years old. She has a bipap machine, cough assist machine, nebulizer all to help her breathe.

Jadelyn has been hospitalized three times due to a mucus plug, each time we would be in the pediatrics unit for weeks till she gets better. She cannot do what her friends do, she does NOT have the freedom. Lately she has been complaining of hip pain due to a dislocated hip she has, as she is fragile...pain management is our option but not sure that's the route we want to pursue being she is so tiny (petite) & how it would actually affect her.

SMA has robbed my daughter’s ability to be mobile and it’s affecting her lifestyle in many ways. She continues to strive, is resilient & determined to live life to the fullest. If there is a cure or anything that my daughter or other children who are struggling & living with this disease can benefit from it is crucial for them to receive it. The FDA has a responsibility & power to push forth the effort by legalizing it and providing our children and family a chance of life. Too many children have died from this horrific disease, how many more???

— Ruthie Panko, mom of Jadelyn, SMA type II

Our precious bundle of joy was born at Jacobi Medical Center in Bronx, NY weighing in at 4 lbs 13 oz, 19 inches long, on January 29th 2004 at 2:15 pm, whom we named Aaron Jai McGirt. He was a perfect baby. When Aaron turned 4 months at his routine doctors’ visit we discussed him not reaching his milestones. The doctor ordered a series of blood tests. As we were waiting for his results from his lab work one early afternoon we noticed that Aaron was having trouble breathing. He started to turn blue, so we called 911 and the ambulance rushed Aaron to S.U.N.Y. Down State Medical Center.
Later that week his genetic testing results were put on rush. One week later we received the results and it was the most terrible news a parent can ever receive. Aaron was diagnosed with Spinal Muscular Atrophy type 1 the most severe. The attending doctor gave Aaron no more than 2 years to live. We were devastated and confused. We didn’t even know what SMA was at the time.

My son’s genetic disorder has impacted our lives tremendously. After his diagnosis at the age of 6 months Aaron had to have surgery and have a tracheotomy and a G-tube placed, and he was put on a ventilator to help him breath. To date we have not been able to bring Aaron home because he requires 24 hour nursing and a handicap accessible home that we do not have to accommodate all of his needs. Due to Aaron’s diagnosis Aaron has no movement and he requires 24 hours ventilation and nursing. He has so much equipment it’s hard for us to fit him and all his equipment in a regular sized apartment.

We would like to one day be able to bring Aaron home. He has been in a hospital setting since he was 5 months old. When Aaron was first diagnosed the doctors told us to enjoy our baby because Aaron would not live past 2 years old, Aaron is turning 12 in January and he is a strong boy and he is OUR SMA HERO! He says that he is happy and wants to keep fighting and that is why today we continue our fight against SMA.

—Myra McGirt, mom of Aaron, SMA type I
Defining Meaningful Change

Meaningful change was relative to current abilities.

LIFE CAN CHANGE ON A DIME. Our family of five was complete with the birth of our 3rd child and first boy in March of 1999. It was clear from the day Griffen was born that he was special. His teachers loved his curiosity and his friends enjoyed his social nature. He participated in every sport he could and loved every minute. By 7 years old he started trying out and making some travel teams. This is when we realized something wasn't right. Griffen fell all the time and his running was getting slower not faster like all the other boys. His coaches were getting frustrated and we were getting scared. We decided to see a neurologist again and more tests were ordered. It was a full year and many invasive tests later that we got the diagnosis, SMA type 3.

By 11, Griffen wasn't able to participate in sports with his school peers any longer, so he joined Wheelchair and Ambulatory Sports! He had a great run for a few years participating in national competitions, but by the time he was 14, he wasn’t able to compete in that world either due to lack of both upper and lower body strength.

Now, Griffen is 16. He is smart as a whip! He started taking AP classes in his sophomore year achieving the highest scores possible and will be taking almost all AP courses this coming junior year. Almost 9 years after diagnosis and there still isn’t anything to preserve his functionality. We have moved to a 1 story home, converted a bathroom, put in ramps and are working on ordering his 2nd wheelchair in 5 years. As a single mom, I am still able to lift him from a chair and the floor, but a hoyer lift will be our next big purchase, followed by a modified van. Griffen isn’t sure how he will make college and driving work for him, but I can tell you WE WILL find a way.

It is hard to watch Griffen lose function when a treatment is so close. Every day the treatment is delayed life gets harder for Griffen. He doesn’t complain and isn’t bitter, none of us are. We still know how fortunate we are that Griffen is in our lives and that his condition (type 3) doesn’t affect his life expectancy, but we also know how much a treatment will mean for him.

—Beth Kingkiner, mom of Griffen, SMA type III
I WAS DIAGNOSED WITH SMA TYPE II AT 18 MONTHS OLD. Despite this, I have gone on to live a fulfilling and successful life. I now live with my beautiful wife, Laura, in Herndon, VA and work as a Software Engineer. When I was first diagnosed it was barely known what caused this disease. I always had hope for a cure, but it was always far in the future. Well, that future is now here. I know now that my generation may be the last to ever be affected by SMA. Having that happen in my lifetime is indescribably amazing. At 27 years old I know that, realistically, I’ll probably never walk. Regardless, having a treatment that could give me more strength would open up so many possibilities that would drastically improve my quality of life. If I could go to the bathroom by myself, roll over in bed, unbuckle my own seat belt, or hold a baby then I could travel anywhere I want, never have to wake up anyone at night for assistance, drive to work by myself, and change my (future) child’s diaper. These may sound like menial tasks to some, but to me it would be a dream come true.

The greatest gift for me that would come from a treatment for SMA is for everyone around me burdened with helping me. More than anything else I would love to be the one helping others.

—Kyle Derkowski, SMA type II

I AM 67 YEARS OLD AND WAS DIAGNOSED WITH SMA TYPE IV ADULT ONSET IN 2007. My symptoms started at the age of 40 but in the past 10 years I have begun to have debilitating symptoms! I cannot climb stairs, walk without a walker and I fall or trip so easily that when I do fall I have no muscle control to catch myself and I cannot get up without assistance. Rising from chairs, toilets and low items is very difficult! I have lost strength in my neck, back, arms, hands, legs and feet. Each year I continue to have less muscle control and realize the difficulties that are ahead for me! I know that this disorder is extremely severe for infants and children and that all research is directed toward a cure for them. But at the same time I really wish that some research could be done for adults that have been diagnosed. SMA has greatly affected my quality of life and my family also! I cannot participate in activities involving my daughters and grandchildren, I cannot take care of my home or yard or travel as I use to do and my relationship with friends has been greatly negatively affected.

The greatest gift for me that would come from a treatment for SMA is for everyone around me burdened with helping me. More than anything else I would love to be the one helping others.

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affected! I’m grateful for the blessings I have had in my life thus far but feel that I could have had many more in my “golden years” if I had not been affected by SMA. A cure would help so many infants, children and hopefully adults in the years to come so I hope the clinical trials will continue and are successful in helping so many people!

—Beverly Smith, SMA type IV

**IMAGINE LIVING WITH A DISEASE THAT SLOWLY PARALYZES YOU.** One that steals all of your muscle strength, your ability to walk stolen before you even had it. Now imagine that you are a bright and social 4 year old boy watching literally every child around you move effortlessly as they play tag, run up a ladder and slide down a slide, play soccer, and ride bikes. This is what children with Spinal Muscular Atrophy (SMA) live with.

Tanner, a bright and happy 4 year old, was diagnosed when he was 20 months old after seeing several doctors to try to figure out why he never started to walk. **At the time of Tanner’s diagnosis I was 7 weeks pregnant with Skyler. We had Skyler tested right after birth and sadly he was also diagnosed with SMA when he was 10 days old.**

Just after Tanner turned 2, despite our constant fight to keep his strength, he lost his ability to crawl. He would slump over on the couch and not be able to sit up again, instead he called to us for help. **He kept getting weaker and we felt helpless.**

Right now the only chance of treatment is to get into a clinical trial. Only the lucky few who happen to be the right age, live in the right distance from a site, and haven’t lost too much strength to SMA are able to participate.

Tanner is one of the lucky ones. **Just before Tanner turned 3 he was accepted in a clinical trial for the drug Isis-SMNrx. Since that time he not only stopped getting weaker, that alone would have made us celebrate, but he started getting stronger.** He started to be able to go from laying down to sitting up on his own. He started crawling again. He started pulling up to his knees and even is able to stand without any support or bracing for a few seconds. His arms are so much stronger. It has been amazing to watch. He has gained so much confidence with gaining strength and getting this treatment has been a huge impact for good in his life. **We would have been thrilled to just have this drug stop SMA from paralyzing him, but we love that it given back so much of his lost strength. All SMA kids need this treatment too.**

**Skyler is so much weaker than his brother.** He has never been able to crawl. Cannot bear any weight
on his legs at all. He can’t stay sitting independently for very long. I wish so much he could have gotten this treatment soon after birth before the symptoms of SMA set in. Time makes all the difference with a disease like SMA. Waiting for treatment versus getting it now is the difference between having a disability and a much, much more severe disability.

It is so easy to feel frustrated right now when there is a treatment out there that works but that only a lucky few have access to. As with any parent who cares about their children, the desperation to keep whatever strength their children have starts to grow. It is hard to stand by and wait when time is not on these kids side.

—Esther Jensen, mom of Tanner and Skylar, SMA type II

Avoiding a decline in function is an important outcome.

MY NAME IS LISA, I’M 52 YEARS OLD AND I HAVE SMA TYPE 3. I have struggled my entire life just to do normal things. I didn’t want SMA to stop me from fulfilling my goals and dreams. Although I had symptoms since about 18 months of age, I was diagnosed at age 20 when I was in college. I decided to put my education on hold and became a mother at age 22 because I was afraid that I would be too weak later on in life. After a divorce, I went back to school to realize my dream of becoming a teacher.

I wanted my children to see strength and determination in me despite watching me fall and struggle to walk. My son learned how to pick me up off the floor when he was 9 years old. My children watched me receive my BA in speech pathology and later on my Master’s degree. I worked for 14 years as a school based speech therapist. After many falls, I was forced to retire due to a severe knee injury that accelerated my progression. Walking is becoming increasingly difficult for me. I am unable to walk outside of my home independently. I like so many others with SMA are waiting for a treatment so that we can live without losing muscle function. I will be a grandmother soon and hope I have the strength to help care for my grandchild.

—Lisa Batista, SMA type III
SMA IS A CRUEL DISEASE. You watch as your child grows older yet grows weaker. You watch as your child becomes too weak to eat enough and must go under surgery for a g-tube. You watch as their spine grows crooked requiring surgeons to cut them open to put in braces in their back so their bodies don’t collapse and crush their lungs and heart. You watch as muscles waste away and form contractures for your child who should have their whole life ahead of them. You watch and worry as the smallest cold could turn into pneumonia and/or death. But there is hope. Research and understanding of the mechanisms behind SMA has led to advances that will lead into many, myself included, see as an SMA free future.

Every pause, every delay means more and more children with SMA dying when just around the corner a treatment was almost available. I understand that there is a process for reviewing drugs to make sure they are safe and tolerable and that steps must be taken, but there is always room for improvement, there is always a way or consideration or answer that shortens the time from when we discover a drug with the potential to help to when it can be provided to those who need it the most.

—Jason Collier, dad of Jayke, SMA type II

MY SON DYLAN JUST TURNED THREE YEARS OLD. He was diagnosed with SMA type 2 just after his second birthday in July 2014. Dylan has never been able to walk. His strength has decreased significantly in the year since his diagnosis, and continues to decrease. He used to be able to creep a short distance while holding the sofa, but can no longer do so. He also can no longer crawl or push himself up to sitting. I spend much of my time carrying him up and down our stairs, carrying his wheelchair in and out of my van, which is also impacting me physically as my back is now affected.

As a parent, it is heartbreaking to see your child unable to participate in the things that other children can do. Our other children do activities, while Dylan gets left behind. This situation has impacted us financially as well. I have not returned to work as I have decided to stay home with him in order to take him to his physical therapy and occupational therapy sessions. Since we do not qualify as low income, we are not able to receive much assistance with durable medical equipment, except for the limited items that insurance will cover. Everything else we have had to pay out of pocket, so we have had to be very resourceful. We are also concerned because we will need to move...
to a one story home in the future, but cannot afford to buy or rent anything in the area.

Our family has been learning to make the best of the situation, however a cure for SMA for Dylan would make such a difference in our lives. To know that Dylan could enjoy the life of a typical child, or at least an improved, longer life, would be an amazing dream come true.

—Angela Chau, mom of Dylan, SMA type II

**Small changes make a huge difference.**

**OUR ONLY DAUGHTER WAS DIAGNOSED WITH SMA TYPE II.** At the time we thought that was the worst feeling of our life but since we have learned that the worse is seeing her on daily basis while she cannot do a lot of things which we all take for granted in our life. The worse feeling is that she is loosing her ability all the time and there is noting other than a good care plan that we can do for her. It is like the person you love the most, gradually dies in the front of your eyes and it hurts that you can do nothing about it. We desperately need a medicine and we know that the scientific community is getting very close to produce one. In fact there are now agents which give a small or modest improvement. The smallest of improvements mean a lot to us. Even the smallest improvement which would be hopefully a sign of stopping or slowing down degeneration matters to us. **THIS IS WHAT FDA DOES NOT GET, I am afraid...We are desperate and fear that FDA does not understand our situation at all.**

—Hassan Sobati, dad of Saina, SMA type II

**SOFIA, LIKE MOST OTHER CHILDREN WITH SPINAL MUSCULAR ATROPHY, WAS BORN SEEMINGLY HEALTHY.** She battles SMA type 1. **SMA, a disease we had never even heard of until diagnosis, is now the ruler of our lives.** It dictates where we can go, when or whether can
take Sofia out, who comes into our home, and causes constant emotional torment. Germs, once a mere annoyance, are now like monsters that we constantly fear will steal our baby from us. Last year we nearly realized that fear when Sofia fell ill. We were advised to take Sofia to the emergency room for chest X-rays. (Later) She was confirmed to have contracted RSV. We lived day to day for those first two weeks wondering if Sofia would ever come home. Her left lung collapsed three times and her right one once during that time. Once she improved, extubation was unsuccessful, so we chose for her to have tracheotomy surgery. It took an additional three weeks for recovery, weaning off narcotics, training for us, and home ventilator trials. After that hospital stay, life has never been the same for Sofia and us as a family. **We live in unending fear for her life and grief at each thought of a future without her smile, whether it’s because she loses the ability to do it or leaves us forever.** We now have nurses in our home 24 hours per day, so we no longer have privacy as a family. Sofia’s room has become a supply closet rather than a bedroom since her bed doesn’t fit in her room with all her equipment and supplies.

> *We live in unending fear for her life and grief at each thought of a future without her smile, whether it’s because she loses the ability to do it or leaves us forever.*

Our living room looks like a hospital room since that is where Sofia spends all her time. Our home has a revolving door for nurses, respiratory therapists, occupational therapists, physical therapists, and orthotists. It takes tremendous effort to take Sofia anywhere if we dare even brave public places with her. It takes up to an hour just to gather and load her “ICU” into our vehicle, which is not equipped for such a large volume of things. **My dream is to see my girl breathe on her own, hold her head up, sit upright, and eat, preferably using her own hands. To most it doesn’t seem like much, but it would mean the world to us!**

—Courtney Roeckel, mom of Sofia, SMA type I

**MY DAUGHTER, SARA ROSE GREENE, HAS SMA 1 AND IS ALMOST 13 YEARS OLD.** She is vent dependent, g-tube fed, and needs full assistance with every aspect of her life. We are so excited for all these promising clinical trials to help the new SMA patients as soon as possible. We are also very eager to try any treatment that may help our daughter improve with any aspect of her life... breathing, swallowing, talking, movement. Each and every day is a blessing and each and every day
we don’t know if it will be the last. Please be kind and gracious to the SMA Warriors and give us hope and options as fast as possible.

—Christy Greene, mom of Sara Rose, SMA type I

MY NOW 2 YEAR OLD DAUGHTER NEOMI RONEN WAS A HALF YEAR AGO DIAGNOSED WITH SMA TYPE II. Not a second goes by that we wish there would be a treatment for this terrible disease. I think about it all the time and I follow all the news. It’s so devastating to hear that your child is diagnosed with a terrible disease, which she got from her parents, which is progressive and which there is no cure for yet. I know we would survive this terrible disease when there would be a medicine that would stop the progression. You can live a happy life without the functions of your legs, it’s not ideal and I really wish there would be a cure, but for now I just pray and hope that the medicines that stop this progression will come soon. Without a medicine fast they lose all of their functions. It starts with the legs, then the arms, then breathing difficulty and the big chance to die from it because of an increased chance of long infection. We live in fear every day for losing her and for seeing her get worse instead of better. I saw a film taken from a child with SMA type II. When she was 2 year’s old she crawled and could play with dolls, she could not stand. Now 20 years later she can only move two fingers of one arm. Her mama carries her into bed. When her noses itching, she has to call her mother to help her, because she cannot move her arms. This is not a life for children. Children with SMA are so smart and communicative, they understand everything, they are a great addition to this world.

—Bellina Ronen, mom of Neomi, SMA type II

OUR ONLY CHILD GABRIEL IS SUFFERING FROM SMA. Initially we struggled for many years and visited over 7 different doctors and specialists in various cities and states to find the causes of our child weakness and inability to perform as a normal child of his age. He was three years old at that time. We spent almost two years before he was diagnosed with SMA.

For many days and nights during many years we have endured the extremely painful reality of not being able to provide help for our son. The emotional wound in our family is constantly refreshed by the crude reality to face the fact that other children do not want to play with him, that he spends
his lunch breaks at school alone wondering and watching other kids play sports and enjoy normal activities, that he is not invited to other children’s homes, that he watches through a window the neighbors’ children ride bicycles or play basketball on the street, that he painfully struggles to climb stairs. Simple things in life and normal basic activities for a child of Gabriel’s age were far reaching for him.

Two years ago, when Gabriel was 11, he was accepted to participate in a clinical trial Isis-SMNRx. Since then, progressively, our son is showing clear signs of improvement, which we had never seen on his first 11 years of age. He does not fall anymore at school or at home, he is capable of climbing stairs, still with difficulty, but with more strength. The impact on his self-esteem and re-gained confidence is tangible; he has performed as an A student since 2013, won first prize in science fair competition at his school for two consecutive years, and was nominated for state science competition in 2014. *His energy level allows him to ride his bicycle, and for the first time in his life, this summer he has been able to spend a few afternoons riding his bike with the neighbors’ children, still at a low pace, but manageable for him. That truly made my wife and I happily cry.*

Despite having grown in height and gained normal weight, Gabriel maintains performing on our own benchmark at home (walking around a delimited perimeter inside our house for a specified time, and we have been doing it for the last three years) We always thank God for this! *We believe that these improvements are worth so much for our family, and that they could do the same for other families suffering from SMA.*

"We believe that these improvements are worth so much for our family, and that they could do the same for other families suffering from SMA."

other families suffering from SMA. They have opened a window of hope for Gabriel and provided us with the energy bust that we badly needed to continue facing our challenges. We pray every day that the research and clinical trials move faster and efficiently because there are thousands of children and families like ours that endure the indescribable reality of pain and hopelessness of seeing the suffering, physical and emotional, of the ones we love the most: our sons and daughters.

—Hugo Cordova, dad of Gabriel, SMA type III
HI I’M ELIZABETH AND I’M A MOTHER OF TWIN GIRLS WITH SMA TYPE 1. Mia & Maya my beautiful angels they are 18th months old. I will never regret to start this daily battled for my girls against an illness that I hope one day close to come a cure can be found to give the innocent children a better quality life.

How is a regular day for a mom whose twins both have SMA type 1? Well, not easy! Mia & Maya both had a tracheostomy and are vent dependent 24/7, G-tube for feeding and also daily respiratory treatments.

Mucus plugs in their airway can happen at any time. We always need to be prepare with cough assist and oxygen if need it. We need to monitor the girls all the time and make sure their oxygen saturations stay above 91% and heart rate below 180. We do trach care everyday after their baths. Every Wednesday we change their trachs and every three months their G-tube bottom.

All mentioned above is not even half of our job as a parent. I did not mention all the stuffs and equipment that we need to prepare for a simple doctor’s appointment or to take them outside. I can keep going on and on with all the work, struggle, frustration and worries that we have to go through everyday, but I have faith that all the therapies and trails to find the cure will succeed if they get approved by the FDA. All the SMA community needs a little of hope for our kids, they deserve a better quality of life and free will just like any other healthy living being.

—Elizabeth Villa, mom of Mia & Maya, SMA type I

I AM A 36 YEAR OLD MOTHER OF 3 WITH SMA TYPE 3. My sister who is now 38 also has SMA type 3. I have a 15 year old son and 2 daughters 4 and 8. When we were young, I was stronger then my sister as she was wheelchair bound from the age of 7 and I am still able to walk a little and use a mobility scooter.

I can remember since I am little how everything was always a struggle. I was not able to keep up with kids at school, my schedule had to be revised to where I wouldn't have to do too many steps or long halls, had help carrying my books. I missed class trips, events in the school, and all the fun stuff we love as kids. Even doing everyday things that people take for granted like getting out of bed, getting off a chair, using the bathroom is a daily struggle.

I always wanted to be a mother and I wouldn't let SMA stop me from doing that too. I have worsened a lot since then as pregnancy was very hard but always try to keep going and not give up. SMA not only causes weakness and fatigue it also causes anxiety with all the what if’s.
Now that I am 36 and have 3 kids my main goal in life is to stay strong for them. It’s a continued struggle as it was when I was younger now just in a different way. Having the strength to get them ready for school, take them to events and all the day to day motherly duties are always a challenge. The SMA not only effects me but it effects my family too. The symptoms I have now with SMA is severe fatigue, loss of stamina, back pain, muscle tightness, neck pain, headaches, anxiety. I can’t tell you enough how much a treatment is needed not only for me but for our families too. This is a terrible disease that slowly steals all your abilities and we need to stop it!

—Woman with SMA type III

“WHY DON’T MY LEGS WORK?” This is a question that a 4 year old should not have to ask. After her 4 year annual checkup with vaccinations, she asked “Will those shots help my legs to work?” This is a question that I wish I did not have to come up with an answer for.

My name is Keanna Nichols, and my 4 year daughter, Brooklyn, has Spinal Muscular Atrophy Type II. Brooklyn was diagnosed a week after her 1st birthday. We feel blessed that we are now surrounded by good doctors, therapist, and family but we are still faced with many challenges. Her inability to walk is the least of our concerns. Our main concerns are respiratory, as those muscles are weak.

She will be unable to attend school on a full-time basis because she tires so easily. We have to give her body time to recoup and rest, or she will get sick. Intellectually she is smart as a whip and very bright, however she is unable to write and color like other 4 year olds because of her low muscle tone. We have to be very careful of where and how we sit her as she easily looses balance and falls over. We are blessed to not be faced with this, but many SMA patients have feeding tubes and breathing tubes. SMA patients are very bright and they deserve a chance to experience the things that “typical” children experience. SMA does not have to be a death sentence, but we need the opportunity to give our children the drugs that can help.

—Keanna Nichols, mom of Brooklyn, SMA type II
About Cure SMA

Cure SMA is dedicated to the treatment and cure of spinal muscular atrophy (SMA) – a disease that takes away a person’s ability to walk, eat, or breathe. It is the number one genetic cause of death for infants.

Since 1984, we’ve directed and invested in comprehensive research that has shaped the scientific community’s understanding of SMA. We are currently on the verge of breakthroughs in treatment that will strengthen our children’s bodies, extend life, and lead to a cure.

We have deep expertise in every aspect of SMA – from the day-to-day realities to the nuances of care options – and until we have a cure, we’ll do everything we can to support children and families affected by the disease.

Learn more about how you can help us reach a treatment and cure at cureSMA.org.

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