A guide to SMA

Stories from New Zealanders living with spinal muscular atrophy

Muscular Dystrophy New Zealand

Contents

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Introduction

Kia ora and welcome.

This resource has been compiled by members of the New Zealand SMA community to help others gain an understanding of spinal muscular atrophy also known as SMA.

It contains information about SMA, as well as a collection of stories that describe life with SMA from a range of perspectives. By sharing these stories, members of the New Zealand SMA community want to ensure that people who receive a new diagnosis feel a sense of belonging to a loyal and compassionate peer network, and that health professionals, policy makers and the community at large will understand more about this rare condition. Together we can reduce the isolation and fear that can come from have a rare condition.

MDANZ is one of a number of organisations that work alongside individuals, families and whānau affected by SMA. We hope you find this resource helpful and would be happy to provide any further information or assistance. You can contact a member of our team by calling 0800 800 337, emailing info@mda.org.nz or find out more about our services by visiting www.mda.org.nz.

Thank you to all who have contributed to this resource.
Ngā mihi nui.

Ronelle Baker
MDANZ Chief Executive and power chair user

About MDANZ

The Muscular Dystrophy Association of New Zealand Inc. (MDANZ) is a not for profit incorporated society that began in the late 1950's as a support group for families affected by muscular dystrophy. Since then, MDANZ has broadened its scope and now provides research funding, practical support, advocacy and specialised information for 60 different neuromuscular conditions.

Our logo is a person shown in the form of DNA. The double helix represents the genetic component to many of our conditions and acknowledges the whakapapa or family histories, which are woven through the stories of our members. Our governance structure ensures leadership of the organisation by individuals or family members with lived experience of a neuromuscular condition.

We have four regional branches that are supported by the national office based in Auckland. Our organisation is a registered charity and we rely almost entirely on voluntary donations from the general public, trusts and other businesses/organisations to continue our work.

Spinal muscular atrophy (SMA)

Understanding this variable genetic disorder

Spinal muscular atrophy (SMA) affects children and adults from all ethnic backgrounds from all around the world. It's a genetic disorder caused by a mistake in a gene called survival motor neuron (*SMN1*).

This mistake leads to insufficient levels of survival motor neuron protein, which then leads to loss of spinal motor neurons also known as anterior horn cells.

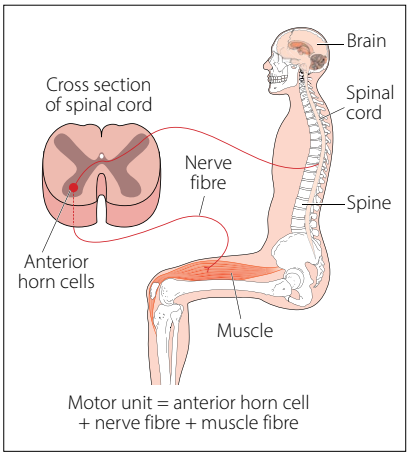
Messages from the nerve cells in the brain (called upper motor neurons) instructing muscles to contract or move are transmitted to nerve cells in the brain stem and spinal cord (called lower motor neurons, including the anterior horn cells), and from there to specific muscles.

In SMA the loss of anterior horn cells means that the nerve signal never reach the muscle, which causes the muscle to waste away (atrophy) as it is never instructed to be used. Like any muscle that doesn't get a work-out it becomes weaker and smaller.

The muscles affected are skeletal muscles and are used for activities such as crawling, walking, sitting up, and controlling head movement. The muscles used for breathing and swallowing can also be affected.

The incidence of SMA in New Zealand is currently unknown but approximately 1 in 6,000 to 1 in 10,000 babies born go on to develop SMA.

Based on clinical features there are four different types of SMA numbered type 1 through to type 4



Motor unit = anterior horn cell + nerve fibre + muscle fibre

but this is really a continual spectrum ranging from severely affected to mildly affected. Very severely affected babies that die before or at birth are sometimes called SMA type 0.

The different types of SMA are defined by the best motor function achieved by that individual – for that reason it can be difficult to predict in an infant what type of SMA they may have as they may not yet be old enough to attain motor milestones such as sitting, standing and walking.

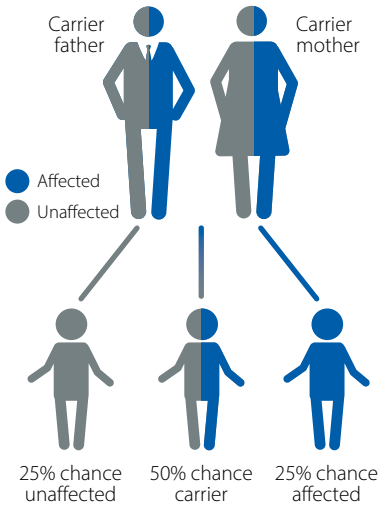
Some people differentiate SMA type 3, which can be very variable in its presentation, into type 3a – onset before 3 years of age and 3b onset after 3 years of age.

Level of SMN protein heavily influences the type of SMA. It is primarily controlled by the SMN1 gene but also influenced by the number of copies of SMN2 gene as SMN2 produces a small amount of functional protein.

Mistakes in both alleles of SMN1 causes SMA and the number of copies of SMN2 influence the severity of the condition, although SMN2 is not the only gene that influences what SMA looks like. The SMN2 gene makes some SMN protein but most of what it makes is non-functional and is discarded by the cell. The small amount of functional SMN that SMN2 produces means that the greater the number of SMN2 copies a person has the less severely affected they will be. The extra amounts of survival motor neuron protein produced by the three or more extra copies of the less efficient SMN2 gene has a protective effect and the severity of the condition is reduced.

Inheritance Pattern

SMA is inherited in an **autosomal recessive pattern** meaning that an affected person has inherited a faulty SMN gene from each of their parents. Both alleles of the *SMN1* gene have mutations that affect the production of SMN protein. The parents of an individual with an autosomal recessive condition usually each carry one copy of the mutated gene, hence they are



When two carrier parents have a child, there are three possible outcomes. With each pregnancy, there is a 25% chance that the child will be affected.

called carriers. Carriers typically do not show signs and symptoms of the condition as they each still have one functional gene, which is sufficient to avoid symptoms. Two carrier parents have a 1 in 4 chance (25%) in each pregnancy of having a child affected with the condition. In rare cases (approximately 2%) people with the condition have a new defect in one of their genes. In this situation only one parent is a carrier and the other defect occurred randomly within the individual. This is called a de novo mutation (new and not inherited).

Diagnosis of SMA

Spinal muscular atrophy (SMA) is suspected in individuals who have;

- A history of motor difficulties
- Evidence of motor unit disease on physical examination
- Electromyography (EMG) records the electrical activity from the brain and/or spinal cord to a peripheral nerve in the arms or legs that controls muscles during contraction and at rest and nerve conduction velocity studies (NCS), which measure electrical energy by assessing the nerve's ability to send a signal.
- Mutation detected in the SMN1 gene (this can be carried out via a blood test or saliva sample) which indicates whether there is a deletion of the SMN1 gene. This test identifies at least 95 percent of SMA.

Features of SMA

Type 1 spinal muscular atrophy (also called Werdnig-Hoffman disease) is a severe form of SMA that is evident soon after birth and results in a rapid decline and death within two years. Affected infants may initially meet developmental milestones but then quickly decline and fail to meet motor milestones although they remain bright and have expressive faces. They have poor muscle tone (hypotonia) and most are unable to support their head or sit unassisted due to the severe muscle weakness. Babies with SMA type 1 have respiratory difficulties and usually require ventilation. They often have swallowing problems that may lead to feeding difficulties, choking or gagging. Other features can include small local involuntary contractions (fasiculations) of the tongue, mild contractures often at the knees and an absence of tendon reflexes.

Type 2 spinal muscular atrophy is characterised by muscle weakness that develops in children between ages 6 and 12 months. Children with type 2 can sit without support, although they may need help getting to a seated position but they never achieve unaided walking. Fasiculations or trembling of the tongue and finger trembling is almost invariably present and approximately 70% of individuals have an absence of tendon reflexes.

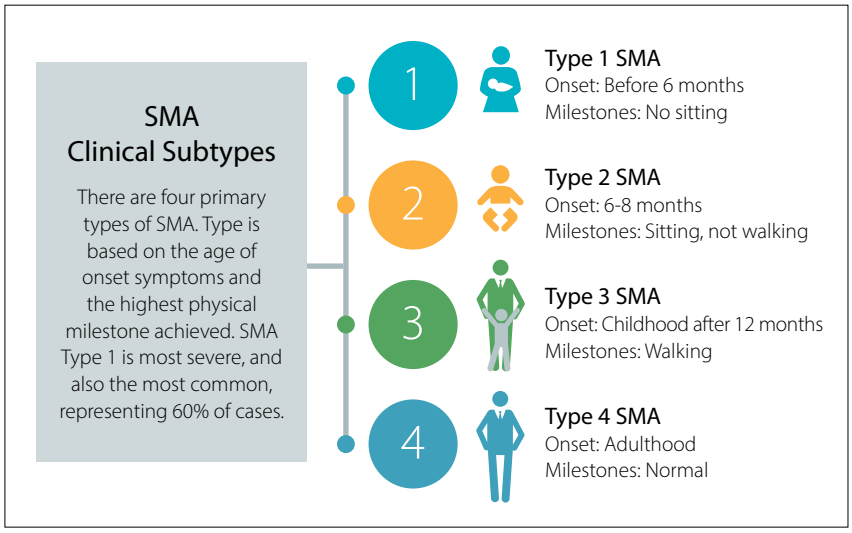
Type 3 spinal muscular atrophy (also called Kugelberg-Welander disease or juvenile type) has milder features that typically develop between early childhood, after ten months of age, and adolescence. Individuals with type 3 spinal muscular atrophy achieve walking but may lose this ability over time. Type 3 is characterised by proximal limb weakness with the legs more severely affected than the arms.

Type 4 spinal muscular atrophy or adult onset spinal muscular atrophy is a relatively mild condition with affected individuals usually experience mild to moderate muscle weakness, tremor, twitching, or mild breathing problems with the onset of these signs and symptoms not occurring until adulthood. Typically, only muscles close to the center of the body (proximal muscles), such as the upper arms and legs, are affected in type 4 spinal muscular atrophy.

Management of SMA

Treatment for SMA involves multi-disciplinary management of the symptoms and prevention of complications.

Poor weight gain, sleep difficulties, pneumonia, scoliosis, and joint contractures are common complications and management revolves around preventing or treatment of these.



Physiotherapy, occupational therapy, and rehabilitation may help to improve posture, prevent joint immobility, and slow muscle weakness and atrophy. Stretching and strengthening exercises may help reduce spasticity, increase range of motion, and keeps circulation flowing.

Assistive devices such as supports or braces, orthotics and wheelchairs may helpful in maintaining mobility.

Good nutrition and a balanced diet are essential to maintaining weight. Chewing and/ or swallowing difficulties can be addressed with input from speech language therapy and dietetics. When nutrition is a concern in SMA, placement of a naso-gastric feeding tube or gastrostomy (PEG) may be appropriate.

It is vital that respiratory problems are well-managed. As respiratory function deteriorates, ventilation, via tracheotomy or non-invasive intermittent positive-pressure breathing device, is effective.

Weakened muscles may lead to curvature (scoliosis) of the spine. Surgery for scoliosis in individuals with SMA type 2 and SMA type 3 can help maintain adequate respiratory function.

Muscle relaxants such as baclofen, tizanidine, and the benzodiazepines may reduce spasticity. Botulinum toxin may be used to treat jaw spasms or drooling.

Regular evaluation by a multi-disciplinary team every six months, or more frequently for children who are weak, to assess nutritional state, respiratory function, and orthopaedic status (spine, hips, and joint range of motion) is recommended.

Genetic counselling is available to families who have a diagnosis of SMA. This service provides information, helps families understand inheritance patterns and what this means in their family, as well as enabling people to make informed family-planning decisions.

Treatments for SMA

There is currently a lot of positive development into effective treatments for SMA, however treatment is not currently available in New Zealand.

Potential treatments tend to target one of the following four approaches:

- Replacing or correcting the faulty SMN1 gene (e.g. AVXS 101 gene therapy)
- Regulation of the low functioning SMN2 copy gene (e.g. Spinraza)
- Neuro protection of the motor neurons affected by loss of SMN protein
- Preventing muscle degeneration and/or the loss of muscle function in SMA.

The current treatment available in some countries is Spinraza (also known as Nusinersen) which has very strict assessment criteria. This treatment is administered by intrathecal injection into the spine and has a strong evidence base for effectiveness against symptom progression. Spinraza works at the gene level, but is not 'gene therapy' as it does not alter the DNA. A person receiving Spinraza therefore requires regular dosing for the treatment to keep working. Spinraza is an antisense oligonucleotide (ASO) designed to increase production of the full-length SMN protein, which is critical for the maintenance of motor neurons.

Other treatments are on the horizon, those currently being trialled include:

- AVXS-101 a gene therapy where a viral vector is used to integrate a working copy of the damaged gene into the motor neurones.
- Risdiplam an investigational SMN2 splicing modifier that is given daily by mouth or by g-tube as a liquid and distributes widely throughout the body. Risdiplam has been designed to help the SMN2 gene produce more SMN protein.

Gavin and Belinda Old have big hopes for daughters Ivy and Olive.

Finding our voice

"Everyone's journey is different," say Gavin and Belinda Old. "But the human spirit is so resilient. We can keep going as long as there's hope. Don't give up and don't take no for an answer."

When Belinda and Gavin had their first daughter, they were like any other first time parents, but as Ivy got older, they began to suspect something wasn't right. At 11 months, Ivy seemed more interested in observing others play than participating, and the family started to worry. At first they were told there was nothing wrong. "People kept telling us she was within the normal range." But when Ivy turned two , and the family had just expanded to include new baby daughter Olive, doctors delivered the bad news. Ivy had SMA. Further testing revealed that the new baby also had the condition.

"It was a knife to the heart," Belinda recalls, and there was limited information provided along with the diagnosis. "We were just trying to put together the picture of where we were heading. The information was not friendly."

"The goal of the first period is to look after yourself and your family," Gavin says. "You have to look after yourself so you can look after your family. It's like the oxygen mask on an airplane."

Family has been an important part of their coping strategies, as has having up to date information from medical and support people. The hope that keeps them going gets transferred at each step of the journey. The biggest hope for now is that the girls will not be held back by accessibility issues. Daycare staff have been wonderful at keeping Ivy involved with the other kids and taking part in normal activities.

"One of the hardest things is watching Ivy struggle with her body not doing what her mind wants," says Belinda. "She says her favourite things are running and jumping but she actually can't do those things. The thought of possibly losing

the ability to paint and draw is so hard but we also have so much joy every day. We measure things in inch-stones, not mile-stones."

"It really makes you focus and look at life a different way," Gavin says. "It resets your priorities."

They say they are grateful for the disability and SMA communities who have made a huge difference to their lives. They hope they will be able to pay it forward for others who go through a similar experience and help improve things for the future.

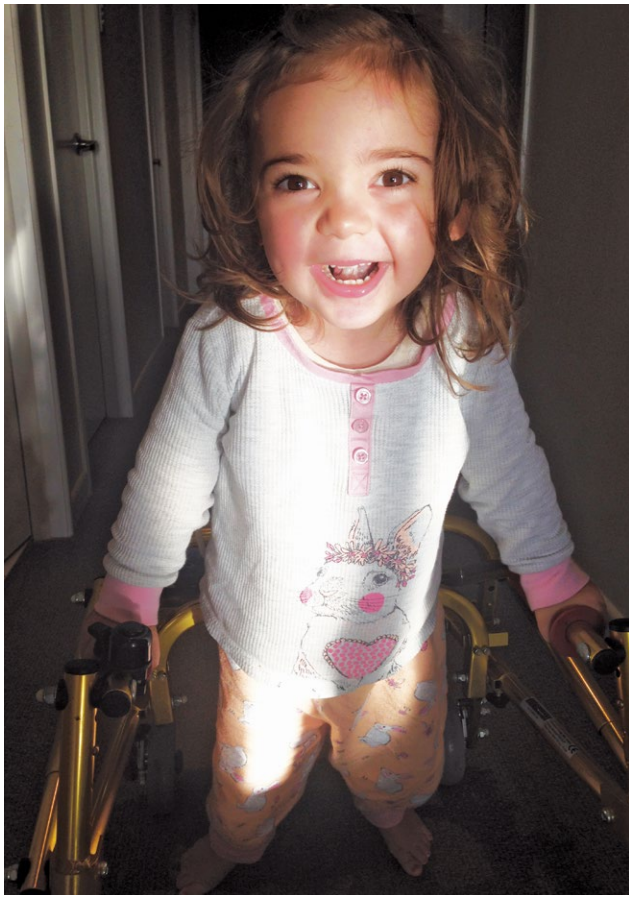
For Gavin, looking at the future is a mixed experience. "It's hard not knowing where the journey will take you. You can't prepare for it. But you always have to be optimistic. Biotechnology is moving forward in leaps and bounds. I hope Biotech companies keep investing in solutions for all rare conditions. It's great to know there's someone out there working on this."

"Singly, our voice is a drop in the ocean, but collectively we make tidal waves."

The family recently moved to Australia to be closer to Belinda's family and in an attempt to get quicker access to new treatments. While they miss New Zealand, they think all of Australiasia needs to focus on accessibility both in the community and in terms of treatments.

"Early diagnosis and early treatment are so important. They help the families get the ball rolling and make a big difference to the outcomes. The benefit over time of treating early is huge in so many areas. It's not about what the treatment costs – it's about what these people can contribute over their lives. There's an impact on society by not investing in treatments. We need to move quickly."

It is that awareness of the importance of keeping up with developments that keeps them involved with organisations like Muscular Dystrophy Association. When asked what important things they have learned from their experiences, they highlight the need to work together to improve things for everyone with SMA. "Singly, our voice is a drop in the ocean, but collectively we make tidal waves."



Top; Olive as a baby. Above; Ivy's walker gives her extra stability.

Colin Cockburn and his sister, Jenny McDonald, enjoying a trip on Lake Taupo.

Overcoming the odds

An incorrect diagnosis when he was a child gave Colin Cockburn a limited lifespan, once that was corrected he determined to make the most of the extra years that have come his way.

Colin Cockburn has always been a movie buff. He has spent many hours watching and enjoying movies in his local theatre and seeing how the audience reacted to the protagonists as they overcome impossible odds. He made good use of this expertise as the manager of a video rental store for some years, gauging which movies would be a hit when they finally made it to tape. Colin himself, has overcome what many would have deemed, more difficult odds than most movie heroes would ever face.

Back in the 1960's, it was noticed that 5-year-old Colin wasn't running like the other children at school. The doctors investigated and believed he had Duchenne muscular dystrophy. His parents were told he would be dead by age 20 and were devastated by the news.

But that didn't stop Colin. While physical activity became more and more difficult, upon leaving school he started his own signwriting business. He began using a wheelchair at age 18 (late for someone with Duchenne in those days) but kept working. Recalling that time, he says, "Around age 25 I thought, I don't feel like I'm going to die tomorrow so maybe the diagnosis is wrong." He was seen by another specialist who rediagnosed him with SMA type 3 and was told to expect a much longer lifespan.

"Suddenly there was a future to plan for after all," he says. He'd never heard of SMA before but he connected with the Muscular Dystrophy Association for support and information. Colin set about making the most of the unexpected lifespan.

Now in his 60's, Colin is retired. He has carers to help him get up, shower, and prepare his food as he can't do these

"Suddenly there was a future to plan for after all".

physical activities himself. His mind, however, is as sharp as always and his days are filled with social contribution, as he is on a wide range of community committees, disability focus groups, a men's social group, Probus, and more. He's an active member of his church, playing a role in services and managing building maintenance. He regularly enters submissions into the Council's Long Term Plans.

Clearly, Colin loves being involved in his community, and provides more than many non-disabled people would, yet he has found it difficult at times. He recalls having his carer hours cut in the 90s and having to deal with 24 pages of questions every year to maintain the level of assistance he needed, despite the clear knowledge that his condition was progressive and not improving. He's noticed the system change for the positive in recent years.

"Since the UN passed the bill of Disability Rights, people have become more aware and it made me more aware. People's attitudes have slowly improved." He says working with the Council changed at that point too. An accessibility committee was established and he's proud of the work they've been able to do, getting an audit of the business district and bringing about changes. Colin thinks a big part of what's needed to create change, is for people to overcome the interpersonal barriers between the able bodied, and those with disabilities. "You have to get to know people and let them get to know you. People are a marvellous asset. Mix with them. It's important for them to see you as a person and know what you're all about."

Attitudes toward disability are not the only thing Colin has seen change over the years. "Technology has changed over time wonderfully," he says. Colin's first electric wheelchair was essentially a manual chair with a motor on each wheel powered by a car battery. He says it had a high risk of flipping over when in use. These days, he has one that is reliable, stable, with added features like leg raising and recline, and a clear indication of how much battery power is left. He says the innovations have been brilliant. Even using a dictation program for his computer rather than having to type is an advancement that has made life much easier for Colin.

One frustration that lingers around the technological advancements is the disparity in funding for it. Colin is frustrated that people whose disability comes from a genetic or medical condition are so much less able to access funding and support than those who gain their disability through an accident and are taken care of by ACC. "Treat people with long term chronic conditions exactly the same as ACC people," is his message for government. "That's the benchmark you should aim for. So there's access to everything they need."

Access to treatment is another important message, and Colin describes the advances of recent new drug therapy as; "Marvellous for young people. I've never been one for getting into miracle cures but this seems to be the most positive new drug therapy and scientific treatment advance in my lifetime." He thinks perhaps the developments might be too late to make a big difference for him, but if given the chance he would try it. "If I could stop the decline of my fingers, that would be marvellous."

After a long life with SMA, Colin has a strong message for those just coming into the journey. "Don't live your life for your disability. Live it for your life. Get schooling, get equipped. "

There's a new movie theatre being built in Taupo and Colin's input is helping to make sure it's accessible. You'll see him there as regular as always, doing what he loves. Because no matter what difficulties might get in your way, he says, "There's always a solution."

Colin enjoyed the company of friends visiting from Colorado USA last year.

Anna and Heath Sutherland.
Photo credit: RDP/Stephen Parker.

Information is power

The Sutherland family continually seek out accurate and up-to-date information so they can advocate for their son to have the best quality of life and medical care.

Anger can be a powerful motivator. For busy mum Anna Sutherland, this is one of the drivers for ensuring she is doing everything she can for her son Heath, following a traumatic and insensitive diagnosis of SMA type 1 at 15 months.

Becoming a strong advocate has been a necessity for Anna, as their diagnosis experience left her and husband Jono poorly prepared with inaccurate information and many unanswered questions. Turning to Google to fill in information gaps was terrifying, as the couple were confronted with the headline "*deadliest genetic disease in children*". Anna describes this as feeling like a 'death sentence', and she was determined to research, network with others and resource herself with as much information as she could, to help Heath, and other families, who were coming to terms with SMA.

Unsurprisingly, Anna now cautions other parents away from Google, recommending SMA support groups on Facebook as a great alternative. "Learning from others with lived experience has been the best source of support, and I feel good knowing I am making a contribution by also sharing information with group members," she says.

Jono and Anna live in Rotorua and have recently moved away from their farm lifestyle to ensure Heath, now two years old, has better access to the community and medical care. Older brothers Jayden and Roman are protective of their little brother and work hard to include him. They made sure he was introduced to the other children who live in the cul-de-sac near their new home.

"It was amazing to see all of the children circled around Heath who was strapped into his trolley, while the boys

explained to the others how to safely pull him around the cul-de-sac. They are very intuitive and helpful," says Anna.

Describing Jono as the 'fun' parent, Anna feels they balance each other out. Jono keeps her grounded and encourages typical family life, while she coordinates medical appointments and filters her world through an SMA lens. Living on one income has a financial impact, but they both know Anna is meeting Heath's needs by being at home and that the sacrifice is worth it.

Recently attending the Cure SMA Conference in Texas, Anna was moved by the adults who shared their stories of what it was like growing up with SMA and why it was important their parents just let them be and do ordinary things, even if it was scary or hard. This changed her view and Anna is now more intent on letting Heath just be himself, and a typical two year old. She feels this approach has helped him with social confidence.

Heath loves his art and is a budding builder. He carefully thinks through his building projects, grouping animals, people and parking cars in the garage. "No one is allowed to help, or touch his blocks," says Anna.

Anna and Jono have high aspirations for their son's future, and want Heath to succeed in whatever he chooses to do. They are realistic that there are physical barriers and potentially serious medical issues that come with SMA, and their recent experience with Heath's hospitalisation and life threatening respiratory issues has brought that reality home.

Heath is now doing better and will soon be seen at Starship Children's Hospital for a sleep study to assess his breathing. They will also be exploring whether to have a PEG inserted to help him maintain and gain weight. This is a scary time for the family, but they will get through it together and with the support of whanau and friends around them.

In a world with endless possibilities, Anna and Jono feel more content knowing they are arming themselves with information and advocating for their son to have the best quality of life and medical care. Treatment is one part of that journey. They are optimistic about access to treatment such as Spinraza, and having this option as early as possible is important for Heath to have the best chance for a more ordinary life, with less challenges.

Anna and Jono have high aspirations for their son's future, and want Heath to succeed in whatever he chooses to do.


Photo credit: Lisa Quirk.

Above: The Sutherland family loves spending time together. Heath during his stay in Starship.

Max, Hayden, Stella, Emma and Harry Beswick.

Growing our family

For the Beswick family, preimplantation genetic diagnosis (or PGD) provided a way to expand their family without the fear that another child would have to deal with SMA.

When her first baby was born, Emily Beswick had never even heard of SMA.

"We noticed a few things that made us a bit concerned, but were told not to worry." Then Emily's ill grandmother spent time watching baby Stella and was convinced something was wrong. By the time the family received a diagnosis, however, Emily was already 40 weeks pregnant with their second child.

"The doctor told us she'd be lucky to make it out of childhood and you need to think about an end of life plan. There was no hope in that appointment."

It was a terrifying experience – not just because of what they'd been told about Stella's prognosis, but also because of what it meant for the new baby due to be born at any moment. There would need to be more testing done to discover if the family had, not one but two children with the condition.

Emily remembers that her grandparents were a huge help during the next few months and by chance, her midwife had previously lost a child to SMA and knew exactly what they were facing. Not everyone did. Emily continued going to her mothers' coffee group and although they were aware of Stella's diagnosis, it was never mentioned. At times, the family felt very alone.

The initial prognosis that Stella wouldn't make it out of childhood proved wrong, but for a long time that worry stayed with the family. Emily would often sneak into Stella's room at night to check if she was breathing. Eventually they learned what was normal for Stella and life carried on, but there have been numerous scares

along the way. A curved spine and respiratory weakness contribute to a high risk for Stella with anaesthetics and in 2014 she spent two weeks in hospital with pneumonia with a temperature of 40 degrees and a partially collapsed lung. Her respiratory health is the biggest concern for the family.

"People think 'it's terrible you're in a wheelchair' but we're fine with the wheelchair. We're actually concerned about lungs," Emily says.

Stella carries on regardless, dealing with medical issues and social injustices with positivity and stoicism. For Emily, the hardest part is the thousand little heartbreaks she feels from seeing her daughter excluded from every day activities like a Kapa Haka performance she'd been rehearsing with the group, only to be left off stage at the crucial moment with no way to get her wheelchair in place with the others. Stella is an inspiration. She took spinal surgery in her stride and doesn't seem to notice rudeness in others when directed at her condition.

"Some people can cope and some can't," says Emily. "It has created rifts but our friends and family are wonderful. We're part of a team. We love the MDA camps, meeting other people who are living with conditions, and the lovely medical people to look after Stella."

New treatments bring the family a kind of quiet hope for the future but it's hard when the cost is prohibitive. "We always thought we would sell the house for a treatment if it came to that but even that isn't enough. It's hard to think she could be getting the benefit of a new drug if we were overseas."

The family hopes that pharmaceutical companies and governments can speed up the process and want them to understand that delays in approving and funding treatments have a massive cost in terms of quality of life. Stella has limited use of her hands now and the idea that she could eventually lose even that is very upsetting. "I'm not expecting that she will get up and walk," says Emily, "but to keep her lungs working and whatever movement she has now would be wonderful."

While tests showed the baby Emily was pregnant with when they received Stella's diagnosis is a carrier and therefore won't manifest the disease, the near miss was a terrifying experience the family didn't want to repeat. They wanted another child but the one in four possibility of that child having SMA was a haunting reality that seemed too much to deal with. Fortunately, a then relatively new procedure came to the rescue.

PGD is a way for parents who have a risk of passing on a significantly impactful genetic disorder to their children to avoid doing so. It involves doing IVF with the extra step of testing the resulting embryos before selecting one to implant that does not have the faulty gene.

Emily remembers this as an emotional and difficult decision and some extended family members were against it. The idea of selecting against a child like Stella was hard to imagine, she says. "But then so was having to think, could we handle losing two children when we're already at risk of losing one? Or can we manage with two children in wheelchairs?"

In the end, they went ahead with the procedure and only one embryo survived so the choice was made for them. The one embryo was a carrier but would not have the condition. The pregnancy went well and resulted in another healthy sibling for Stella.

When it comes to PGD and early testing opportunities, Emily has a simple message: "Don't judge if we do and don't judge if we don't." These are difficult decisions that no one can fully understand until they are in the situation. She is grateful that the options exist and hopes the future brings more opportunities to improve the lives of those dealing with SMA.

Hayden and Stella enjoy some summer fun.

Kerry was very proud to be a groomsman at Rebecca's wedding.

Changing the future for others

Four years after his death, special memories of Kerry Hills are still very strong for his family and friends.

Rebecca Barbarich remembers her brother Kerry Hills as a man who loved life, family, was very humorous and determined to achieve whatever it was he set his sights on. "He kept our family connected. He was my best friend, we were super close sharing everything," she says.

Rebecca was three years older than Kerry and when her little brother was born, no one realised anything was wrong at first. However around the age he should have been learning to crawl, concerns began to form because he wasn't able to do what other children his age could. He was originally diagnosed with muscular dystrophy, but this was later revised in teenage years to spinal muscular atrophy (SMA). Rebecca made a point of learning about her brother's condition and even gave a presentation to her school class, as part of a research project on the topic.

"We didn't have the supports there are today," she says, thinking back to those early years. "Dad made him a trolley and we used that in place of a wheelchair."

This family trait of ingenuity and ability to think outside the box was something Kerry had plenty of. He was a determined young man who always found a way to do the things he wanted in life. And Rebecca found herself helping him achieve his goals whether she agreed with them or not. ""He was the most driven person I knew and never let his disability stop him. He didn't do self pity or take charity. He was always eager to help others."

When Kerry decided he wanted to live away from the family home, Rebecca and other family members were worried for him, but Kerry made it happen. He got a job and used his income and funding to hire support and structure

them in the way he needed. He was active and lived life to the fullest, planning a cruise for his 30th birthday. Kerry played wheelchair football, travelling to Australia on a few occasions to represent New Zealand in the Power Blacks team and returned with numerous medals. He was the president of the Auckland Powerchair Football club and dedicated a lot of his personal time to managing the team.

Despite his independence, Rebecca never lost her big-sisterly protectiveness towards Kerry. They were both vigilant about keeping people who were sick away from him, aware that illness was a greater risk for him than for others. Unfortunately the day came when all their precautions failed and Kerry was admitted to hospital with what would turn out to be swine flu.

"It was awful and scary," Rebecca remembers. "They stuck needles and pipes into him and told us he was going to die, but he hung on. He fought better than an able-bodied person would have."

The family rallied and Rebecca hardly left her brother's side for two weeks. They considered their role as both carers and advocates vital as, even in intensive care, they felt staff didn't seem to fully understand the needs of Kerry's SMA-affected body. Rebecca felt that perhaps some medical staff misjudged Kerry's quality of life and the reason he fought so hard to stay alive. "It felt like they saw him as a frail shell not the strong person he was. It was horrible as a family to be so helpless."

Eventually, Kerry lost his battle with the illness. "I felt heartbroken and alone. I used to talk to him every day and we'd go out on weekends, shopping and taking his dog for walks. Losing him was a big loss." Thinking about that moment still brings tears to Rebecca's eyes. She leaned on friends and family and her husband to get through that dark time. "You have to try not to let the grief consume you," she says. "Because you could. But Kerry wouldn't want that."

At his funeral, Rebecca was amazed at how many people her brother's life had touched. His determination and willingness to help others had influenced many people both in and out of the disability community. "He achieved a lot."

Years later, life is good for Rebecca but she still misses her brother. "After eight years, we've finally had a baby and it is heartbreaking that Kerry's not here. They would have loved each other." Rebecca and her husband decided to have genetic testing before starting a family and discovered that while Rebecca is a carrier, her husband is not. There is a risk their child could be a carrier, but no chance of her actually having SMA. "I didn't worry about it. It isn't going to affect my baby and even if it had, it wasn't a burden to me," says Rebecca.

"He was the most driven person I knew and never let his disability stop him. He didn't do self pity or take charity."

She thinks Kerry would have been excited to see the developments in treatment for SMA. "Kerry always wanted to be a guinea pig for treatments. He wanted to help change the future for others. He knew it wouldn't necessarily make his life longer but would help others." She thinks funding for treatments is a "no brainer" as it brings hope.

For those who are waiting for those treatments, she says, "Don't give up. Don't let the disability restrict you. It's just a shell. It's what's inside that counts. You'll have lots of good times." And she knows because she shared many good times with her brother. "Kerry never let his shell stop him."

Kerry's first fishing trip. He was super proud of himself for catching and wheeling in his first snapper using an electric rod and reel.

Tania and Regan Woodmass, with son Lincoln.
Photo credit: NZ Herald/Alan Gibson.

Finding a family of support

Receiving their son's diagnosis was isolating and frightening, but since then Tania and Regan have discovered a community that's like a family.

The internet has been both a blessing and a curse for Tania and Regan Woodmass. A blessing as a source of information and support, but a curse in terms of giving them a harsh introduction to the condition they'd never heard of until it invaded their lives in the form of their son's diagnosis.

The family had never heard of SMA but were gradually becoming concerned about Lincoln's development. Staff at his preschool said he was falling a lot and they felt something was wrong. Tania and Regan took videos of Lincoln and took him to a doctor for advice. Initially they were referred to physiotherapy, but this didn't seem to help so they went for a second opinion and the videos were sent to a specialist.

Receiving the diagnosis was devastating. They thought Lincoln was being tested for SMA just to eliminate the worst outcome. "And then we were told that worst outcome was reality." There was little information for the family so they had to search for their own and this was a heart-breaking experience. "We did our own homework on the internet and that was bad as you get the worst-case scenarios. It wasn't until later that we found out many SMA kids are so smart and do more than many able-bodied people do."

They felt isolated at first and are incredibly grateful for family support during that initial time. Gradually, however, the internet began to be helpful after all. They found an SMA NZ group on Facebook and learned that the Muscular Dystrophy Association of New Zealand supports those living with spinal muscular atrophy as well. Finding that

"It opened our eyes to disability and having empathy. We see people doing great things despite disabilities. It doesn't have to mean you're stuck in a box."

support meant a whole new community of people who understood and could help them learn and deal with the situation.

Finding what they refer to as the "amazing SMA family" has made a massive difference to their lives. "It means you're not alone," they say. "We can offer support and we all have common ground that we're linked with."

"It opened our eyes to disability and having empathy. We see people doing great things despite disabilities. It doesn't have to mean you're stuck in a box." This was an important realisation for them about their son. "Lincoln can be great and he doesn't have to be put aside by society."

The kindness of people and their communities has touched them in many ways. Work contacts and friends went out of their way to raise funds to help. While it can be hard to watch Lincoln play with his peers and not be able to keep up, Tania says she often sees his friends and cousins slow down so Lincoln can catch up, or even win, a race and they cheer him on. Those moments of kindness mean the world to a three year old who needs help with many physical tasks others his age can manage, but always tries nonetheless.

"Get the right information and the right people," is the couple's message to anyone coming into the world of SMA. "You're not alone. Don't give up. It's not the end."

Getting connected to the right people and information has meant the family is now part of a focus group on SMA issues, and they have their fingers on the pulse of new developments. They're very excited by new treatments but frustrated by the length of time it takes to make those treatments available.

"Treatments need to be at the top of the cliff, not the bottom."

They want early testing and fast treatment. Particularly when it comes to approving and funding new treatments. "Our children get weaker as you delay. How can you do that? Stopping this disease means supporting productive members of society. We've come such a long way. Now we just have to hurry up!"

Lincoln loves playing with his friends and cousins, and tries hard to keep up. Photo credit: NZ Herald/Alan Gibson

On holiday in Los Angeles.

Appreciating the little things

Brent Walker doesn't let his condition stop him living life, but gets frustrated when others can't see past the stereotypes.

"Having SMA has taught me perseverance," Brent Walker says. "It also teaches you to appreciate the little things. You miss a lot of big things so the little things turn into big things."

Brent was diagnosed as a child and he remembers little about it from those early days. "I just remember being frustrated that my body wouldn't do what I wanted and my legs were sore when I tried to walk." He remembers life being good as a child and being somewhat puzzled to learn about his difference. For Brent, growing up with weak muscles and a wheelchair felt normal and it wasn't until he began to notice the way others behaved around him that he began to understand about being different. "I spent time at school trying to decipher why people were that way to me. It gave me a lot of time with my thoughts and to understand myself."

As he grew up, Brent made sure not to let his condition stop him from living his life. He has travelled extensively around America and Hawaii, works for the Ministry of Social Development, and is studying economics via the internet. He also volunteers on the Muscular Dystrophy Association National Council and the SMA focus group.

Life and travel take a lot of planning to make it work but Brent says it's worth it, especially to have his own place. He hires carers himself and runs his supports like a business to make sure he can live the life he wants. He says it requires him to be good with money, and with people, and to be very organised but it's a necessary evil. The alternative is to give his money to an agency or move into residential care, which he would consider to be "pretty much the same as prison."

He's used to having to plan for every alternative, however, "having SMA affects every aspect of my life. I'm constantly thinking about accessibility and comfort. Going anywhere new is hard. Will there be parking? Is it accessible? Is it raining or crowded?" There are times when outings are just too difficult and he will have to stay home in order to avoid the risk of illness. As his condition has progressed, Brent has to be more and more careful to avoid getting sick.

"It doesn't take much for me to get sick and it takes longer to get over it." Brent is careful to take care of his physical needs like nutrition, staying warm, not getting run down, and using a cough assist machine, but he also considers his mental wellbeing to be a big part of keeping healthy. "Anxiety is exhausting and exhaustion leads to getting sick," he says. "Staying healthy in your head is a massive thing."

"When I was a kid, every second person stared. Now people are much more used to us."

There are some hard times for anyone diagnosed with SMA, but Brent believes the future is looking good in terms of science, accessibility, and society. "When I was a kid, every second person stared. Now people are much more used to us." That said, he still finds it difficult to overcome stereotypes sometimes and thinks people often just assume without taking the time to get to know someone in a wheelchair. He wants people to realise that SMA is "just physical and we're not slow in the head so don't interact with us like that. When I'm in a shop and they just talk to me normally, it means a lot."

It's a little thing but those little things are significant. When it comes to advances in treatment, little delays are incredibly frustrating for Brent. "I was positive about treatment at first but after a year and a half I try not to think about it too much." He keeps himself up to date with the latest developments and is excited to see children who won't have to deal with some of the difficulties he has lived with but, he says, "I don't see why it has to take so long. For such a developed country we don't have any excuse. I understand there's a limited budget but they need to streamline the assessment processes more. I get annoyed that bureaucracy is what's preventing me from improving or stopping my decline."

Brent believes investing in treatments will save thousands in the long-run on equipment and hospital visits and enable people like him to work more and be more productive for society.

"I live a pretty normal life but wish I had more freedom to just be normal and not have to constantly adjust to the progression of the condition."

Brent loves to travel as often as he can.

Tina and Gary have made sure their daughter Logan (now 8) has grown up hearing all about her big sister Tylah and understands her special place in the family. Photo credit: PhotoWorthy Images

Our girl Tylah

Life changed in an instant for Tina and Gary Bell when their baby daughter was diagnosed with a condition they hadn't even heard of.

Simple everyday moments conjure up special memories for Tina Bell. Her baby daughter Tylah is there in the feel of a feather brushing against her skin, the sight of a helium balloon, and the sound of running water signifying the start of the bath-time she adored.

Life is full of unmet milestones, this year would have seen Tylah celebrate her 13th birthday, and Tina and her husband Gary enter a new phase as parents to a teenager.

By the time Tylah was six weeks old, Tina was concerned that she wasn't getting stronger.

"I remember a lady making a comment about how her grandson was always kicking his blankets off, and I suddenly thought; 'You know what? Tylah never kicks her blankets off'."

The realisation started a string of conversations with the Plunket Nurse, midwife, GP, and a couple of specialists. Initially the couple were told Tylah had low muscle volume that would improve with physio, but Tina instinctively felt there was more to it than that.

"I left there and rang my mum and said to her that the doctor was wrong, I just knew it."

More appointments followed and after an agonising five-day wait between specialists, the couple were given the SMA diagnosis.

"Suddenly there's this condition we find out we are carriers of, and we haven't even heard of it. Nobody we knew had heard of SMA – not one single person," says Tina.

"We were basically told to take her home, that there was nothing that could be done."

Tina instantly went into mama bear mode, becoming a passionate advocate for her daughter and everything

she needed. When told physio might be available several months down the track when the condition would be at its worst, she made sure Tylah had access to it right away.

"My attitude was you can get stuffed if you think my daughter is not having the best quality of life possible, I fought for everything that was right for her.

"We fought every day. I swore I could beat it, but I was wrong. They told us she would only live to six months of age, but we got her to 9 months and 19 days."

Although Tina finds it very hard to watch, the family has hours and hours of video footage of Tylah taking part in the things she loved.

"I remember people saying they couldn't believe how much we did with her. We would have done more but SMA robbed her. It robbed her of being able to sit, it robbed her of being able to crawl. We were limited in what we could do, but we found everything we could that would be suitable for a child with limited mobility. I'd lay her down so she was comfortable and give her a paint brush, or we'd have lightweight things to play with like helium balloons or feathers.

"She loved the water, and would cry if she heard the water running and didn't get put in the bath. Her little sister is a water baby too."

Although they never met, eight-year-old Logan knows all the things she has in common with the big sister who came before her, and Tina says her daughter often feels sad she didn't get to meet Tylah.

"They are completely different personalities," says Tina. "Tylah was so placid, Logan is the complete opposite. She certainly makes us smile every day. But she gets sad she never met her sister and cries for her."

Tylah's death plunged Tina into a dark and difficult place and it has taken years to come to terms with.

"People say time heals, it doesn't. You learn to live with it, but the grief is always there."

"We fought every day. I swore I could beat it, but I was wrong. They told us she would only live to six months of age, but we got her to 9 months and 19 days."

Tina struggled with people who either didn't know what to say and ignored her pain completely, or gave unhelpful advice. Friendships and relationships become very strained. After reaching rock bottom, she eventually had counselling, which made an enormous difference.

"A couple of weeks after her 11th anniversary, I was sitting at work and I just felt this darkness come off my shoulders. To the world I often looked happy, and of course, I have my daughter and she makes me happy, but there is still that darkness. Then that one day, I felt it lift, and I was able to take a step forward. It will never completely leave me, but I can move forward – it took 11 years."

Over the years, Tina has spent time with other parents of children with SMA and helped them with the grief process, and the practicalities of getting the best care and knowing the right questions to ask. Tina says everyone's journey with SMA is different, but the support of others who have walked the same path makes all the difference.

She believes there is more information and better support for families now than there was a decade ago, and ongoing research is encouraging. However, news about the development of Spinraza was bittersweet.

"Of course we want it to happen for other people, but it still really hurts. Why couldn't it have happened for us?"

Photos of Tylah are very precious to her family, she shared a special bond with her cousin Maddy (far left).

Sathy and Dharshi make sure their boys participate in everything they want to – cricket is a favourite pastime.

The joy of twins

The Sathiyaseelan family has made a commitment to focussing on the present and giving their boys every opportunity to live life to the fullest.

Twins are an exciting addition to any family, and hearing the news during their routine antenatal scan brought Sathy and Dharshini Sathiyaseelan great joy. Already proud parents of a son, Sathy and Dharshi welcomed twin boys Ashvin and Arvinth in 2006. They were based in Singapore at the time, having moved there from their home country Sri Lanka, for Sathy's project work in the construction industry.

The family moved to New Zealand when the twins were a year old. They were progressing well and began pulling themselves up to stand, but were very wobbly when walking and would fall over backwards, hitting their heads. Seeking advice from their GP, it was noted that twins in general were sometimes slower to walk, and nothing serious was identified.

The head teacher at day care however, observed the boys in a much more active setting and noticed a difference in their gross motor development compared to others their age. Following specialist assessment and genetic testing, both boys were confirmed as having Type 3 SMA when they were two and a half years old.

"I was given information, but the paediatrician told me not to read too much. It was quite scary, because we had never heard of SMA before", says Dharshi.

The knowledge that they were carrying a recessive gene that caused SMA was quite a shock for Sathy and Dharshi, who have no known family history. While it was challenging coming to terms with the initial diagnosis, two things gave them some comfort – the fact that their eldest son was unaffected, and that the twins both had the same condition.

"The boys always have each other and they are never alone in this experience."

Being new to New Zealand, Dharshi was quickly shunted into a learning process as she began to navigate the health system and coordinate multiple appointments for the twins, while Sathy maintained full-time work. She had to get her drivers license in order to maintain her caring role and now proudly drives a modified van that hauls two power chairs – though avoids Auckland's motorways if she can!

An early piece of advice received from Paediatric Neurologist Rakesh Patel was to focus on the present. The family had lots of questions about the future for their sons, and Rakesh was quick to point out that all children are different and the rate of symptom progression would be difficult to predict. The family took this advice and decided to focus on the present and being with their boys each step through the journey.

Ashvin and Arvinth are considered "brainy kids" who are sports mad, and their broad interests have been fostered by school and their busy parents, who ensure their participation in everything possible. Adaptions need to be made at times, for instance the boys will bowl rather than bat during cricket.

Dharshi and Sathy acknowledge that the environment and resources available to support disabled people are better in New Zealand.

"Things are very positive here. Like the support the boys receive to attend a mainstream school. There have been building modifications and they are included in school life, with a teacher aide and therapists who know them from primary school."

"The boys always have each other and they are never alone in this experience."

At times the boys bring up the emotional impact of SMA and this can be a challenge for their parents. Answering the 'why' questions, and making sense of what is happening as symptoms get worse, is never easy. And as with most 12 year olds, Ashvin and Arvinth are independent thinkers and occasionally may not want to ask for help. Dharshi tries to manage these situations with kindness.

The family continues to have a neurological review twice each year and this is where they hear about research and medical updates. They are positive about potential treatments that may become available in the future, to help the twins maintain current function, and prevent symptoms getting worse.

In the meantime, their focus will continue to be on the present and living life to the full.

Ashvin and Arvinth both love sport and enjoyed the chance to meet their cricket heroes.

Being the best me I can be

Living with a type of SMA that isn't immediately obvious, brings its own challenges for Samantha-Rose Williams.

For Samantha-Rose Williams, SMA has been an invisible disability. Diagnosed at age 14 with type 3, she says people often don't understand why she gets so tired easily, or struggles with climbing stairs. She often finds herself in the role of having to educate people about her condition. "I have a friend who's gay and kind of has to come out to people every day," she says. "It's a bit like that. You learn how to explain it quickly. You get used to it."

While most of her friends are quick to accept and understand the limitations, Samantha has had difficulties with others in the past. Sometimes she gets dirty looks when using an accessible carpark, despite having a permit, and she's found some workplaces aren't willing to make the adjustments she needs to give her body a rest and struggle to grasp that she has good and bad days.

She says it's important to learn your limits and be strong about stating them. When going out with friends she will insist on driving rather than walking, or suggest she wait at the cinema while they go to the supermarket for snacks. She knows to pace hersel,f and rely on handrails or another person for support.

"I was initially afraid to tell people but the reception was mostly good. I got to know who my friends really are." She doesn't let it bother her if people drift away because of it as those friendships were likely to fail anyway. "Hey, if you have an issue with this, then neither of us need to bother trying."

Having SMA helps you realise what you want in life, Samantha says, and it works like a sieve for sorting out the important things and people.

She doesn't let her condition stop her from achieving her goals. She's in the final year of her bachelor's degree and is in the process of writing a novel. She loves fashion and romance and writing fun characters, as well as going to the movies and "eating lots of sugar with friends."

Samantha-Rose never misses an opportunity to sing along to High School Musical.

While the word pessimist doesn't spring to mind when you meet Samantha, that's how she describes herself when it comes to future medications. "I don't want to sit around waiting for treatment. I'm planning my life as if SMA will stick around. Treatment would be a dream come true but it doesn't seem realistic for me yet." She knows that as someone with a comparatively mild form of SMA, she may not make the cut for funding for medications as those with type 1 and 2 come first, but she says it's good that there is now a hope for everyone with SMA.

She's also conscious of rare diseases like SMA getting left out of funding entirely. "Really well-known diseases like cancer get a lot of focus and lesser known ones fall under the radar. Just because there are fewer of us doesn't mean we should be ignored."

For now, her advice to others with SMA is to take one day at a time. "When you think about the future and what might happen, that's when it freaks you out. Be easy on yourself. Yes, it will affect you but it isn't all of you."

"I'm just trying to be the best me I can be," she says. "That's all anyone can do."

Looking to the future

Ensuring early diagnosis, access to treatment, and ongoing support

This resource has provided information about SMA and shown a broad range of experiences and perspectives about living with this progressive, often fatal condition.

In New Zealand, our community is united in the shared vision for quality lives, access to good care and effective treatments, and supporting research into potential cures for SMA.

As we look to the future, it is encouraging to see progress being made toward the early diagnosis and better treatment of SMA.

Newborn screening

We are taking steps to have SMA included in the Newborn Metabolic Screening Programme in New Zealand, to ensure early diagnosis and access to treatment intervention as early as possible in a child's life. This offers the best possible health outcomes for the child.

Early diagnosis also enables parents to undertake genetic counselling and family planning before having more children. This facilitates informed choice if they wish to explore the options available to them for having another child not affected by SMA.

Treatments and cures

At present Spinraza is the only current established treatment for SMA and this is now funded in many countries, but not in New Zealand, currently. New Zealand has a current focus on improving access to funded treatment for rare disorders, including SMA. There is reason to be hopeful for the future as a range of new treatments are being developed and trialled.

Collaboration

We are committed to working in collaboration with local and international partners, to advance the united vision for people with SMA to have the best care, and effective treatments. Through our activities, we aim to build social capital, grow our leadership capacity and drive positive change for the New Zealand SMA community. Globally, August is now recognised as SMA Awareness month.

Social support

Our community leaders will continue to create opportunities for individuals, families and whānau to share information, network, and build social resources. SMA is a lifelong journey with many challenges, but it does not have to be a lonely one. Through building a network of support, we can ensure better and timely supports for our community members, helping each other to be resilient and better prepared for the future.

Resources

At MDANZ, we want our resources to be inclusive, inspiring and informative. Together with the SMA community, we will facilitate access to SMA-related resources that can help people in their early journey with a new diagnosis, parents to have conversations with their children about what is happening to them and how they might handle different situations in their childhood lives, and health-related information so people can manage symptoms and maintain their quality of life and well-being.

Muscular Dystrophy
New Zealand

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