Appendix

1. **Living with the Condition**

Example Case Studies

1:1 **Adult Type 2**

You never know what is round the corner, and whilst you hope things will get better, you never dare to think too far ahead or of the 'what-ifs'. There are days when the disease wins or the mental load is too much but, other days when you win (for the first time this week you didn't choke eating dinner, you managed to brush your own hair, you felt less tired at work, you made your children supper without assistance) then, your appreciation of life goes up dramatically. You live for these moments because you had days where you couldn't even do those things. With SMA the small things are BIG things - you appreciate the simple things that others take for granted, and if you should on occasion dare to dream, you wish for more of these simple things or for them to last even just a little bit longer.

1:2 **Child Type 2**

Arthur was diagnosed with SMA type 2 at 18 months old, we had concerns at around 9 months but were told not to worry until 12 months. At 12 months we made an appointment & had a 6 month waiting list to be seen, a few days before the appointment Arthur was admitted to hospital with a virus and the doctors were very concerned about his lack of mobility and floppiness. Shortly after we received the diagnosis.

At that time it was absolutely devastating, reading about SMA we were absolutely broken. We had no idea if we could keep working, if we would lose our house and how were we going to cope watching our son deteriorate before our eyes.

SMA has had a profound effect on our lives as a family. We have to juggle lots of appointments, for check ups, treatment, Physiotherapy, hydrotherapy & Hippotherapy.

We have faced issues with Arthur's nursery & him not being able to be with his peer group due to the class being upstairs and health and safety issues with no 121 support being offered by the council, so he has to stay in the baby class until we can get him into pre-school. This is detrimental to his cognitive and communicational skills as SMA does not affect his intelligence, he has struggled seeing his peers move up to the next class.

Financially it is extremely tough, we have spent a lot of money adapting our house and garden for Arthurs needs. We have also invested in equipment both at home and nursery to aid his progression. We also had to buy a bigger car for his equipment as he is under 3 years old he doesn't qualify for Motability scheme.

Living with a child with SMA is emotionally draining, I have recently contacted the doctor for counselling and treatment for stress and anxiety. It is a constant worry about the future and his independence.

Treatment makes this bearable, of course this isn't the life we would have chosen for Arthur, however the access to treatment has been lifechanging and gives us hope. Arthur initially lost strength before his diagnosis and has gone from only being able to roll & sit without being able to reach out for anything to crawling, standing & walking in his walker. Who knows what the future holds if the drug is approved as he is going from strength to strength. I believe he will be able to walk and gain more independence to do things for himself if the drug is approved.

It is inhumane if these drugs are not approved after the clear benefits this brings. Arthur is gaining strength every day and without this I don't know how I would be able to watch him deteriorate; it just doesn't bear thinking about

1:3 **Adult Type 3**
My name is Steven Jones and I have SMA type 3. I use a wheelchair full time, but live a majority independent life in terms of living on my own, driving, going to work and I currently work in the civil service.

I started Spinraza in the summer of 2022, and this was a really interesting time for me, I've never really seen my disability as something that needs treatment or needs support. But throughout my twenties, and I'm 30 years old now, I had seen not s rapid decline but a very noticeable decline in my abilities. In my early twenties I used to be able to get up, go around the house on my knees if I wanted to, go upstairs, I can't do that now . I also started to find transferring really difficult and really noticed when the height of the bed, chair or toilet was a bit lower, it was really difficult to get back into my wheelchair. That caused me quite a lot of stress and anxiety thinking, if I'm going to public places, what would I be able to do?

Getting treatment was quite a difficult decision for me because I was thinking, do I need it? What will be the side effects of having a lumbar puncture? I went for Spinraza because that provided more certainty because of the data at the time compared to Risdiplam.

So leading up to treatment, we were just coming out of lockdown which I found quite difficult for my body because instead of going to work every day I was sat at home and I think with SMA, if you don't do certain activities you lose the ability to do them. I was really tired every day, I found it really difficult to get out of bed, I didn't really know what my fatigue level would be from day to day, what I wanted to do or what I could do. That provided a lot of uncertainty for me.

I used to avoid going out because I used to be worried about the accessible bathrooms, would things be the right heights? Will I feel really tired? How will that impact on my day?

Once I started Spinraza, things started to look a bit more hopeful, I had a noticeable increase in energy and fatigue levels dropped. Of course that wasn't instant, but looking back compared to what I was like at the end of 2021 start of of 2022, I now have a lot more energy and I'm able to continue to live an independent, fun and happy life. That's been the biggest noticeable change. I haven't noticed the impact of transferring anymore so that would be another noticeable change.

And secondly, a bit of hope, those in the SMA community have to come to terms with change in their disability over their lifetime. I'm expecting, as I get older, my abilities to decease even further and that's quite frightening. I think; What will I be like when I'm 40 years old? 50 years old? Will I be able to work when I need full time care? But what Spinraza does, it helps to stabilise the condition and I've noticed that stabilisation in my condition. That has enabled me to hold a bit of hope for the future, hope that I will be able to live a full life, go on lots of holidays and continue to work, continue my social life and my hobbies and activities. That is invaluable and I think that stabilisation is really the hidden benefit of this medication. Its not about helping me to walk again like I used to in my teenage years, but actually that stabilisation is so valuable to me as it keeps me independent which is actually such a full part of my identity and who I am.

Looking back, I would not have changed my decision to get Spinraza, I find the lumbar puncturescompletley fine, a couple of minor side effects but they are not really noticeable, I

can almost forget about the medication for four months, go back to work or on a holiday, enjoy my life knowing it is coming up in 4 months and have a bit more certainty for the future.

1:4 **Adult type 4**

I was diagnosed approx 5 years ago where I had to leave a job I loved for 25 years as manager of local Airport for Loganair as I couldn't do the hours due to extreme fatigue and experience anxiety which was very new to me being a very calculated person before SMA.

It changed my whole character which I am still trying to manage, after changing jobs 3 times in 5 years desperately trying to find a way to continue working for my family and my self respect, I now work at local bakery, leave home at 4.30 to start 5 on Mon Tue Thurs and Fri, I have to have Wed off, as I have to recharge and always take the weekend off.

I have fallen many times as the condition manifests mainly in right leg but my arms now are getting weaker.
I have to work for my family who are still children, my pet annoyance is web sites terming my type 4 condition as normal and being able to live normal life, from the Inside it is not normal.
I'm always told how rare my condition is and I struggle to find anyone else to talk to, if I got Risdipalm it would totally change my life, give my confidence back and maybe stop further deterioration.

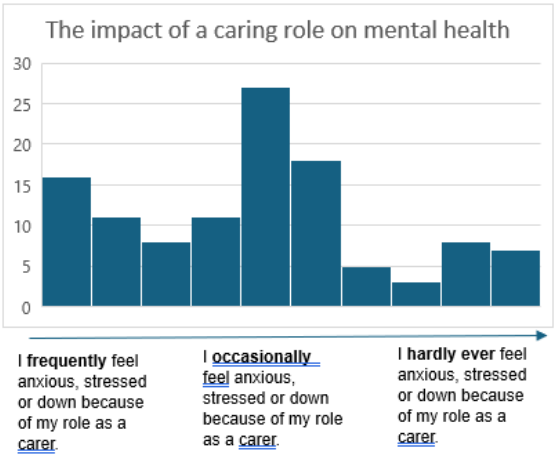
I try to continue working as it is my physiotherapy as well as providing mental and financial help but don't know how long I can carry on.

Desperately looking to be an active member of society as I have always been for myself and my family, I Have developed a hermit personality as you are self protecting which is not living.

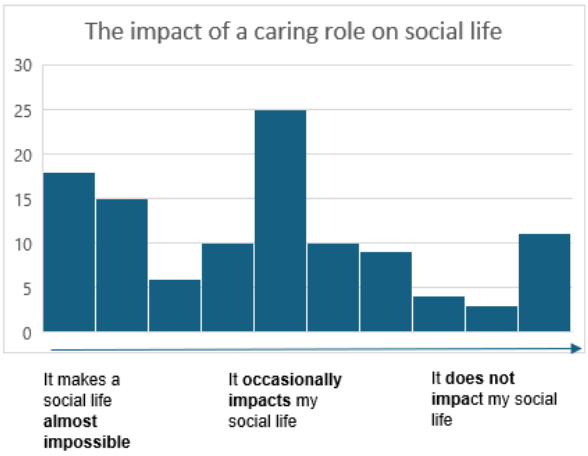
2 The role of a carer

2:1 Quantitative data

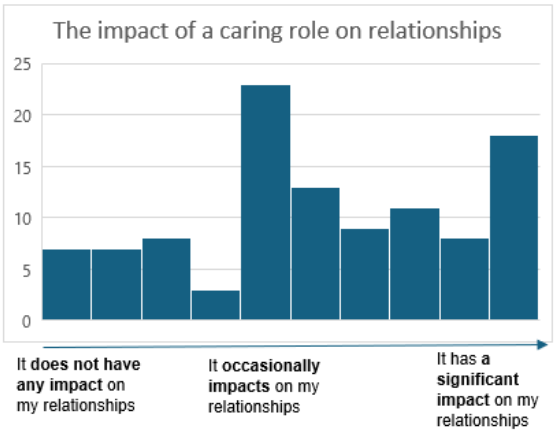
Mental Health and Wellbeing



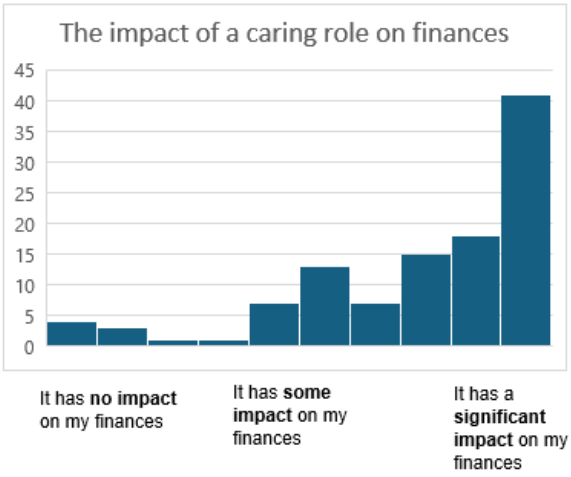
Social Life



Relationships



Finances



Qualitative data

2:2 **Carer Mental Health**

57 free text respondents

Themes

- Anxiety for the future and changing care requirements.
- Guilty that they aren't doing as much as they should to access therapy, equipment and enabling inclusivity. This is intensified by social media enabling comparisons amongst the SMA community.
- Guilty that the genetic condition is the 'fault' of the parent.
- Anxiety that the person they care for might get a virus that makes them very poorly, always being in 'alert mode'.
- Depression having to watch the person they care for suffer through colds or miss out on opportunities due to a lack of inclusion.
- Stressed from having to juggle appointments and fight for services, therapies and equipment.
- Constant fatigue from waking nights.
- Coping with trauma from intensive care admissions.
- Worrying about future access to treatment.
- A lack of mental health services to provide counselling for both carers and those living with SMA

Random selection from the 57 comments

As grandparents and fortunate enough to be able to share the role with maternal grandparents, seeing our grandson wheelchair bound and physically limited makes us feel responsibly guilty, sad and anxious for his future and also for that o fhis parents.

Looking after our son with type 2 (weak) has changed our lives, the constant alert you are on knowing that any illness or cold could have a very damaging effect. Henry has been on treatment for 4 years, it's changed all our lives and lessened the burden on our mental health and anxiety for the future.

I am the Mother of a type 2 little girl. She was born this way so I have adapted to her care needs. Since she's been on treatment I have witnessed her meeting milestones that she has NEVER met before - this gives me confidence & I feel positive about her having a bright

future. If she was not on any treatments I would feel anxious and depressed - especially if I could see her declining. Her day to day care is something I don't choose to feel sad about - I remain positive and love watching her thrive. Her happiness and positive development through treatment has and IS helping me to be a strong, happy positive Mummy.

There are a few mental challenges that come to mind: - the worry/guilt that as a parent I am not doing enough to help her condition - the need to suppress any frustration (with our situation, with her behaviour) to not deflect stress onto her or my son - the worry/guilt that her brother isn't having the same opportunities because of our limitations - general stress of time management around work commitments - stress of single parenting and co-parenting with ex spouse

Massively stressful and exhausting always being the sole carer to someone. You don't get a break or time to yourself. Outside services are limited and a lot of the time we feel very alone.

It's very difficult to be a parent of a child with sma and the role this comes with. Even things like toileting which is impossible in public make me anxious to go to places. Working is difficult and funding everything is hard.

I am, on the whole, ok dealing with the everyday needs of my son but I have always struggled with thinking about what the future holds for me. I am also very tired emotionally & mentally chasing people/organisations/nhs who are involved in our lives.

A caring role is a difficult role for anyone caring for a loved one but as a mother who has cared for 23 years to my daughter with SMA it has had it challenges physically and emotionally in would say my mental health has been affected in many ways at different stages of her life I.e teenage years were a struggle. Now she is an adult my anxiety is more affected due to worrying about her independence and her health treatment had helped to ease these stresses slightly as although not a cure for her SMA I do not worry as much with deterioration of her condition

.2:3 **Carer Social Life**

Random selection from the 53 comments

As a parent to young child, my social life is impacted anyway. However Beatrix does have extra needs as all SMA children do - so my social life does get effected - HOWEVER, when I do get time to socialize I am more mindful to enjoy myself because as the back of my mind I know my child is having treatment & it is a positive impact. I feel if Beatrix was not on any treatment, fear, anxiety would over take me and I wouldn't be able to enjoy to socialize as much.

My wife's bones are weak and she is going on a trial to increase to try and stop breakage. As her hip was broken by the scan team in their mishandling of her. This has increased my freedom 100% as she has had her hip ball removed. She needs her leg moved and the current wheelchair is impossible.so I have had to give up work so financially lost but she has great stress when I leave her now so this has a big impact to my live and wellbeing.

Care requirements mean it is difficult to find someone else who is able to care instead of me but now she is consistently attending school things have improved significantly.

Due to my sons care needs I don't have anyone else who can look after him suitably so I just don't go out

I cannot just go out, it takes planning. My husband and I cannot go away together.

No holidays away for me for respite for 15 years. It is difficult to take any time off from my role

I'm pretty drained and tired most of the time, after a full 12hrs of being present I find it difficult to socialise after work as it's either late or I just need some me time

I practically have no opportunity to find time for myself, my daughter is next to me all the time

Ensuring someone is actually here to cover the shift, worrying about whether everything wok at home, not knowing if you'll be called home for an emergency

No family members know how to look after my child. I don't have a social life to be honest.

I am fortunate enough to have excellent parents and a partner who all support me in being able to maintain some sort of social existence. However, this is still different to that of my friends who have children who don't have sma. Trying to find babysitters or carers outside of your immediate circle is virtually impossible with the higher level of needs required

I can still lead a relatively healthy social life due to family support but I have found it difficult to work with having a daughter with SMA due to doing everything for her down to itching an itch on her head! Although we have help from carers this is just for personal care ie showering and getting dressed the normal day to day tasks are down to myself my husband and her sisters .

2:4 **Carer Finance**

55 free text respondents

Themes

- **Equipment:** Only the most basic needs are covered by the NHS.
- **Therapy:** The NHS does not have the capacity to meet the needs of the SMA community. The lack of physiotherapy support in the adult community is stark. Many families are also paying privately to top up any physiotherapy provision from the NHS.
- **Adaptations:** The accessibility grant system does not always cover everything required by someone living with SMA. For those adults that are working, they are required to self fund a proportion of the costs.
- **Energy:** From extra heating to keep carers warm overnight, to electricity to power ventilators, hoists and power chairs, it is well documented how living with a disability increases energy costs.
- **Cost of living:** Inclusive activities come with a higher price tag. For example, extra rooms have to be booked for carers on holiday, there is not the choice of restaurants and leisure centres that able bodied people have.

Random selection from the 55 comments

I have been unable to work full time and have to pay for private physio and equipment not funded by the NHS

Previously the main earner of the household and now a full time carer. Had to rely on relatives to pay our mortgage. Fundraising has been necessary to ensure our child gets the

correct care to provide things like orthotics and private physiotherapy and other equipment that the nhs have been unable to provide

Due to having to give up work we are down to a one income house now. We don't receive any extra child benefits. We use disability allowance to purchase necessities for my son, pay for private physio, etc.

It does, Electricity bills more than anything, but you do get help with this with the disability elements and i use the DLA towards these bills and fuel for regular appointments

Everything that is needed for a disabled person like equipment etc always is more expensive, holidays are more expensive due to needing to accommodate the disabled person and energy bills are larger with charging of equipment keeping the home warm as my daughter is constantly cold all this on top of all the pressure on my husband to financially support us alone as I am unable to contribute and work and carers allowance for genuine carers is pennies for the work we do ! so yes finances are definitely impacted massively .

Financially it is very expensive to care for someone with a disability. Luckily we are able to work still, however the cost of a suitable home, wheelchairs, equipment, WAV and physio is very very expensive.

As the parent of an adult with SMA living at home, energy consumption is high due to all the equipment used and most days being at home all day.

2:5 **Carer Relationships**

45 free text respondents

Themes

- Sleep deprivation
- Social isolation
- Physical pain (particularly back pain)
- Post-traumatic stress
- Financial difficulties
- General anxieties about the person being cared for and their future

Random selection from the 45 comments

It has changed the marriage between my husband and I to the point where we exist as carers only. We work together to care for our children but are separate at night due to one of us on night shift in another room. We are handling daily stress and sadness about our child and it makes us argue/ feel frustrated with each other.

My husband and I have no time alone together, it's an extraordinary juggling act to be able to spend time with the other two children 1 on 1. It's hard.

It's hard to maintain friendships when you can't go out much. Intimate relationships can be affected due to always being tired or stressed

It has had quite a big effect due to my personal time, I feel there is not a huge amount of work life balance'

SMA affects every single aspect of our marital relationship and I'm constantly doing a 360 risk and safety assessment everywhere we go, which is draining and makes me feel more a parent than a spouse'

Being a carer and going to work and trying to maintain successful/positive relationships can be challenging and, whilst I am fortunate to have a supportive and loving husband and parents, I would say many of my other familial and friend relationships have suffered as a result. This is often through being excluded due to lack of accessibility or not understanding inclusion.

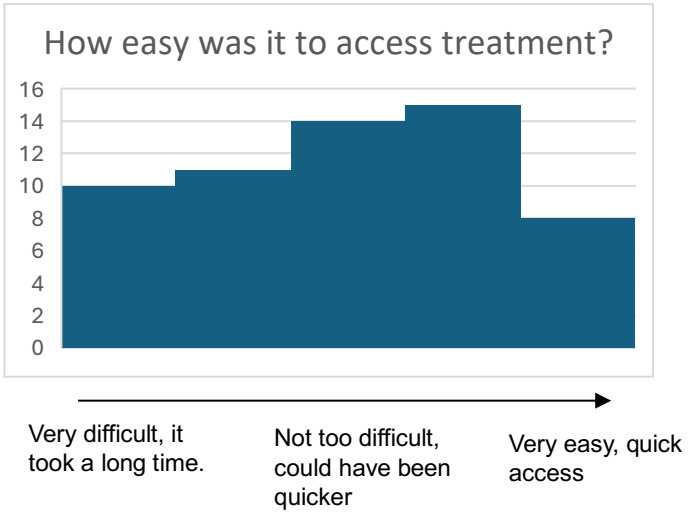
Meeting with friends with children of the same age makes me feel isolated. My child can't play at the park, walk through the woods etc. I therefore avoid meeting up.

Marriage of 25 years ended because caring for a child/young person/adult has a massive impact on relationships. Making sure a younger sibling is able to enjoy life too creates a huge strain and leaves you juggling many balls in the air. Tiredness is overwhelming and leaves you feeling it is too much effort to do anything else but care for your loved one.

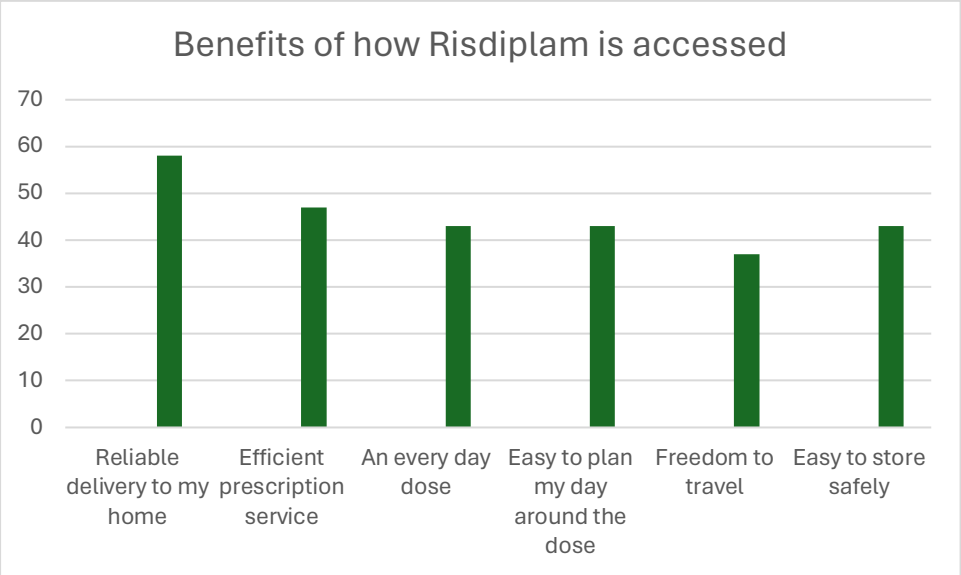
Being a carer role for my daughter has to a certain level effected my marriage - but don't all children. She is able to go to school etc and mastering new strengths and stay in her school. So we manage to keep our marriage sustained. Without treatment, I can only imagine the disease getting worse therefore having more and more impact on my relationships

3. What are patients' and carers' experience of accessing and having these treatments?

3:1 Quantitative Data

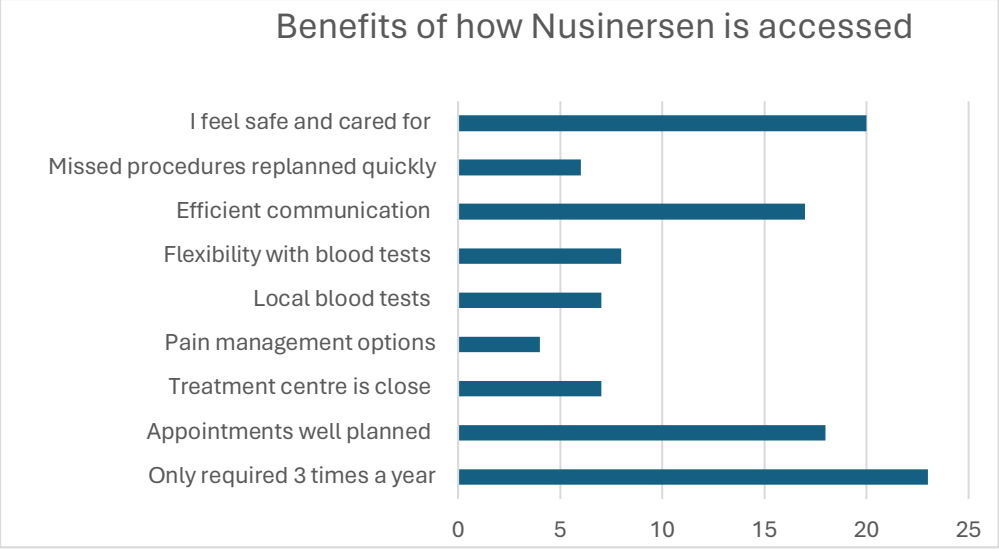


Risdiplam



Most respondents could not name any negatives to the way Risdiplam is accessed

Nusinersen



3:2 Qualitative data from both treatments

Random selection from the 32 responses

Took a long time to be offered MAA. I think because there weren't many people of my age and the hospital wanted to offer the drug simultaneously to a meaningful sample.

I am a Ukrainian refugee and I was surprised that I am eligible to get the treatment without having citizenship. I enquired my GP and he referred me to Oxford's clinic where I did the genetic test and was waiting for its results and doctor's prescription for several months.

We have always had a good experience with accessing treatment.

No negative aspects. Feel blessed to receive life changing treatments

Access to my treatment has always been a positive experience. Staff are fabulous and the care & support is super.

The team are all extremely caring, experienced, and they do their job thoroughly.

My hospital (The National for Neurology) took quite a long time due possibly to the issues of staffing around COVID, to put the treatment programme in place.

Fought with many others in Glasgow and surrounding area to get onto trial then having it approved by Greater Glasgow & Clyde Health Board. We contacted everyone from First Minister Head of NHS, MSP`s, Councillors and were about to go to press when it was approved by NHS to be administered for ever.

3:3 Access when fully ventilated

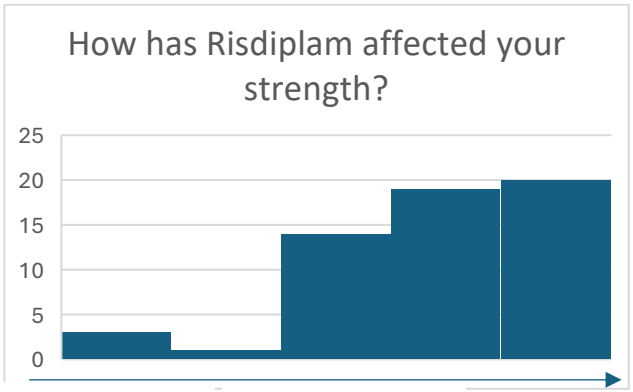
[Our son] was born 9/1/21 and shortly after birth showed signs on SMA, he was rushed to Evelina and intubated early Feb, diagnosed mid Feb. It was agreed Zack would start Spinraza and was given his first dose. I was then told there is a strict criteria in order for him to continue with treatment which included him not being invasively ventilated, so it was unknown whether we could continue with the treatment long term. After 2 doses of Spinraza he was still intubated and I was told we'd possibly have to stop treatment, it went to an MDT and was decided to let him finish the loading doses of Spinraza and then we'd see where we were after there. After the loading doses he was still intubated but gaining some movement (bearing in mind he had little to no movement with a CHOP score of 3)

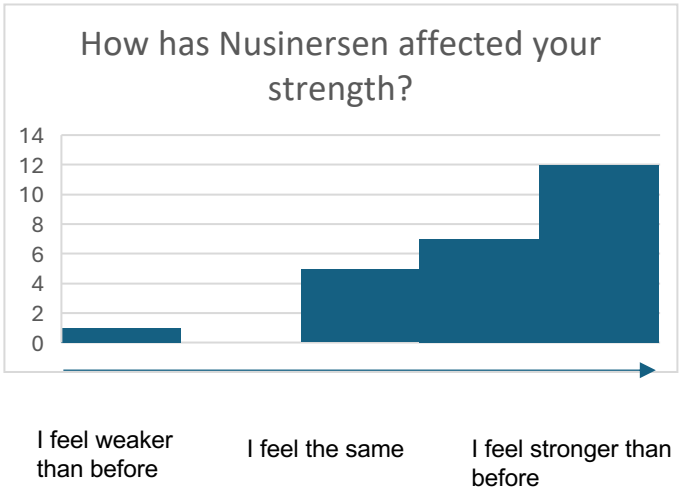
It was then 4 months until he was due another dose and it was rocky as to whether we'd get there but during this time he was extubated onto bipap which initially started at 24 hours a day and working our way up to bigger breaks. He was still barely managing 8 hours off of bipap when to my surprise we were offered Zolgensma.

If he had not finished those loading doses I'm certain he wouldn't be here with us. The criteria currently in place does not give the weakest children a chance to rest and gain the strength needed in order to continue with treatment

4 <u>What do patients and carers think are the advantages of the treatments?</u>

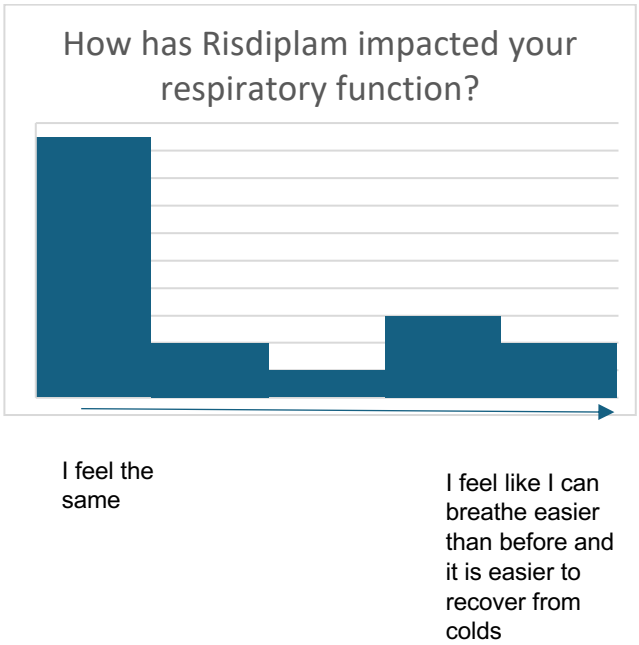
4:1 Strength

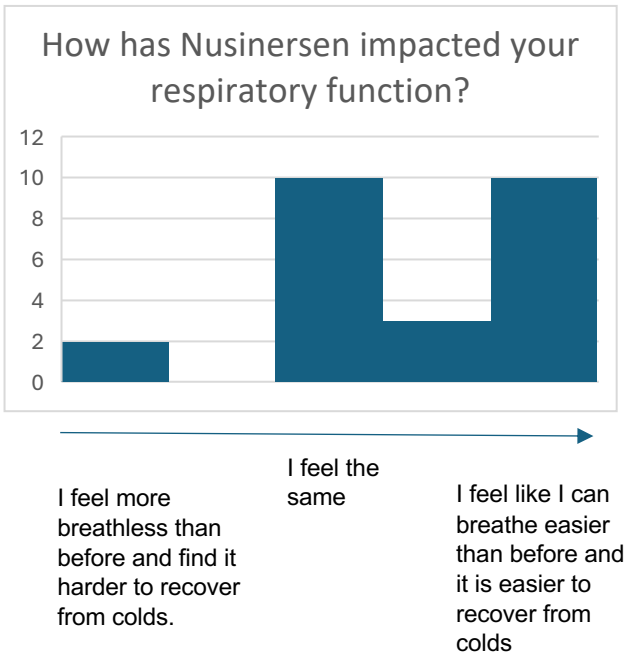




Respiratory Function

Nobody taking Risdiplam reported a deterioration in respiratory function since starting the treatment.

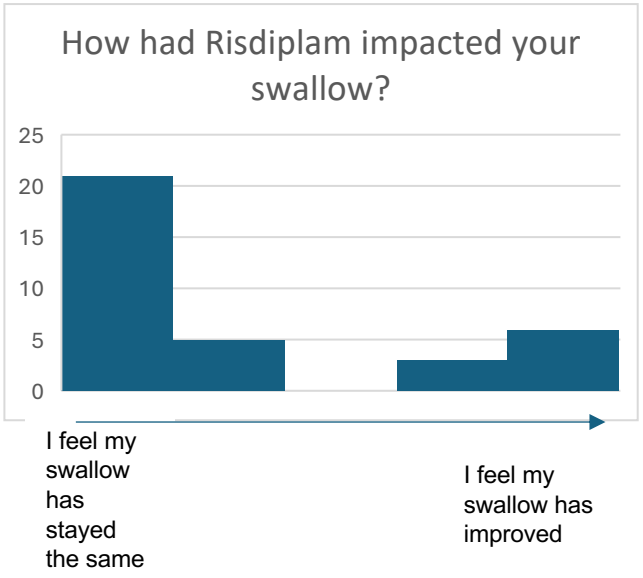


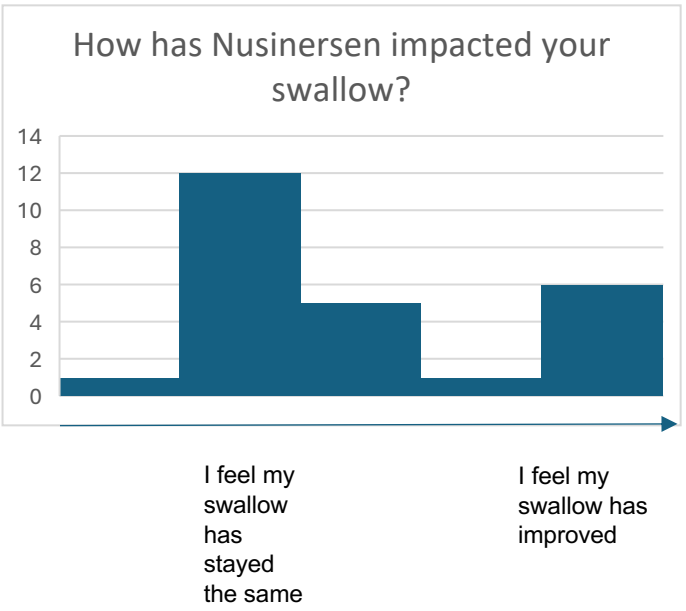


One of the respondents who reported a decline in function, an adult living with SMA type 2, seems to have been an error as they also commented in the free text:

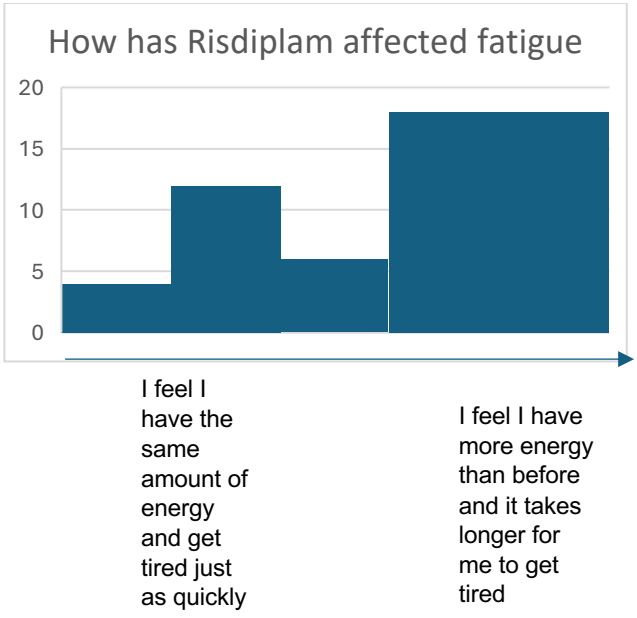
'I used to have morning headaches but these have gone. O2 is much improved was 95, now 98 or 99%. This makes me life significantly better.'

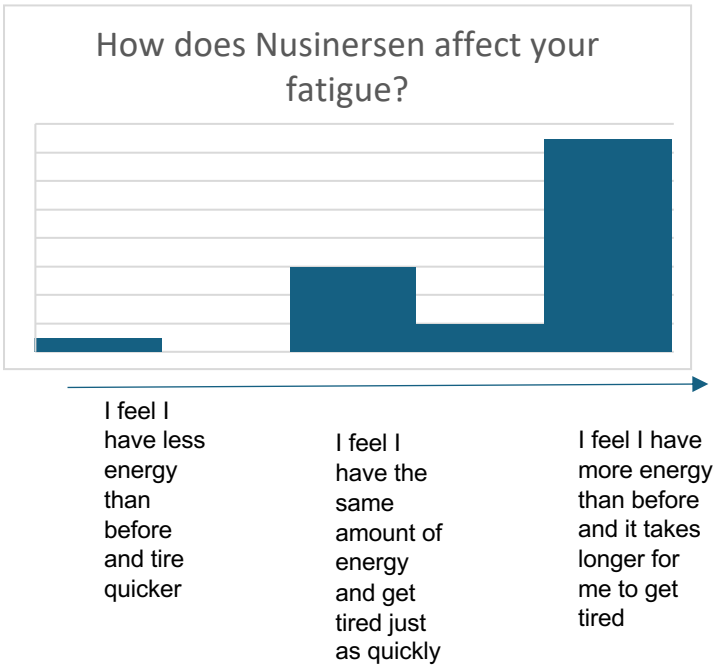
Swallow



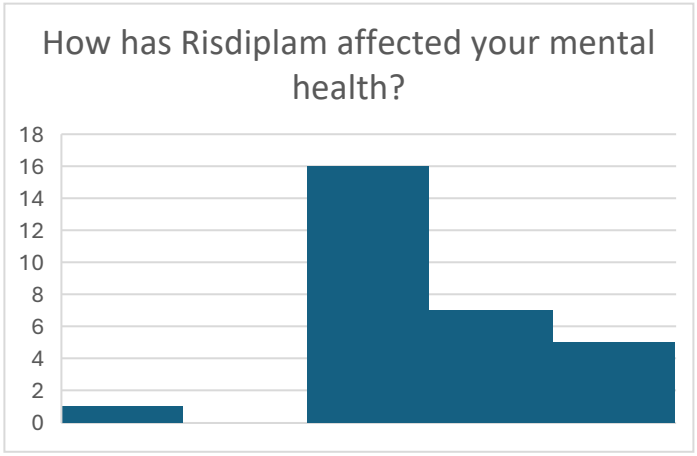


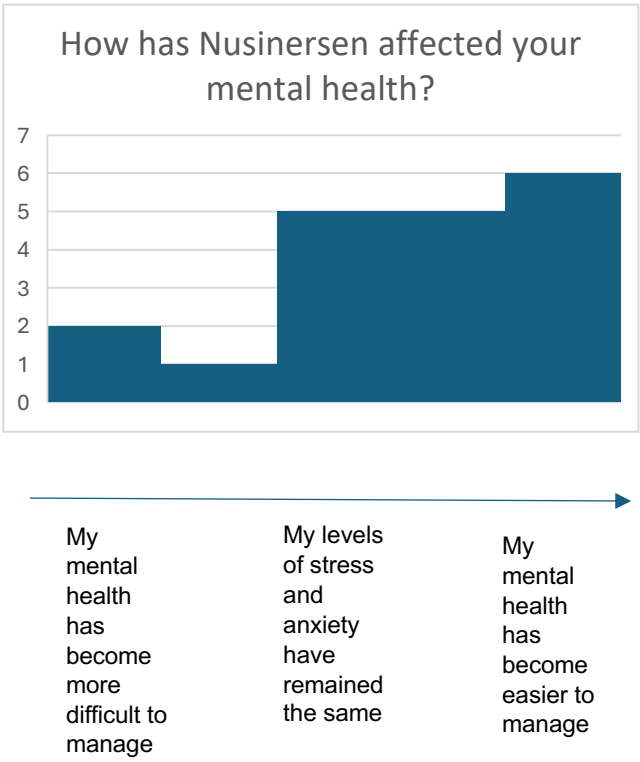
Fatigue



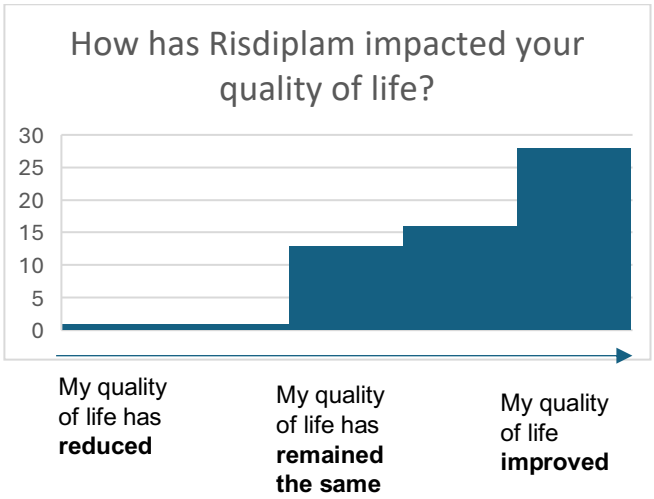


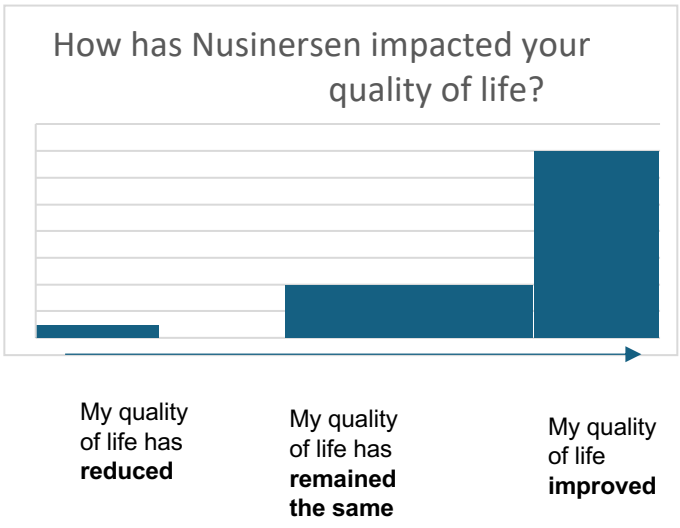
Mental Health



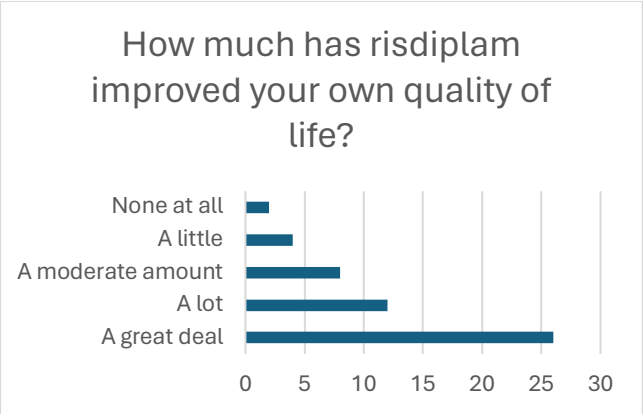


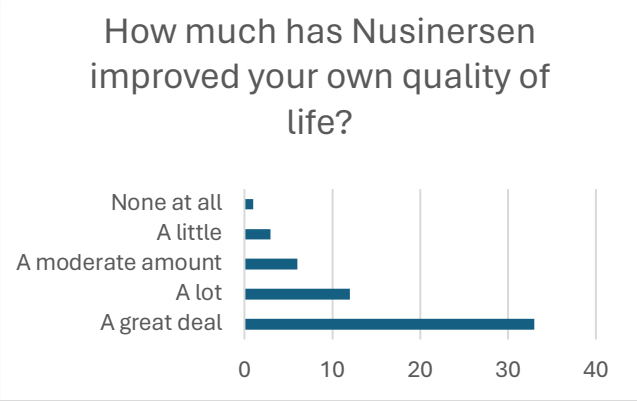
Quality of life





Carer Quality of life





4:2 Treatment outcomes

Strength

'*Better finger strength can type at work with both hands when previously couldn't. Can lift left arm if it falls off my armrest where previously couldn't.'* Adult living with type 2 SMA

'I can now lift my foot off the ground (never before). I can pick up a laptop and move it, without calling my Assistant. Lifting my arms to feed myself. Much, much stronger core strength - can sit up unaided for first time in 20 years' Adult living with type 2 SMA

'I have more energy and can better control my head and neck when driving outside in my wheelchair - this means I can go out without a carer, and even this small gain in strength has given me a huge amount more independence and freedom. I'm a university student, and since taking Risdiplam I've been able to walk around campus with friends, because I've not been worried/needed support for my head control. Before this gain I found it virtually impossible to go anywhere without a carer, and now that I can I've been able to meet new people and make new friends, simply by being able to go from one place to another without help. This small gain has had an immeasurable impact on my life.' Adult living with type 2 SMA

'*When I started [on treatment] I was not expecting any improvements, I just wanted my disease to stop being progressive. I was afraid of the possibility to lose my physical strength because my hands are already pretty weak and losing strength at the point means losing the ability to do my routine completely. But after three months of taking Risdiplam, I felt like I could sit longer, not being tired after the day at the University. And I can spend much more time in the wheelchair without feeling fatigue the next day, so it is therefore me to travel, to study, and now I am even*

having a placement at the Gallery of Contemporary Art as a magazine writer.' Adult living with type 2 SMA

Respiratory

We used to have antiobiotics every winter when he would get periods of illness, but the longer he has been on risdiplam the better he has got and he has not needed any at all and has had no time off school for illnesses this winter.' 'Co child living with type 2 SMA

'I have slight asthma, with my improved diaphragm strength and breathing this has almost 0 effect on my life now whereas before it could be debilitating at times.' Adult living with SMA type 3

Swallow

'*Before taking Risdiplam, I was starting to have some difficulty swallowing certain textures of food and I had to angle my head to the right to be able to swallow food and after taking it for a few months, I noticed that this issue got better and I could swallow any textures however I still have to angle my head to the right to be able to swallow'* Teenager living with type 2 SMA

'I find my swallowing has improved. I think there is still an issue but not as bad and now I can manage it better. I have never eaten quickly anyway but know I couldn`t now for sure. Just take my time and be careful. But definitely never avoid food of any kind (I love it too much). Adult living with type 2 SMA

'I used to suffer from occasional choking which has almost completely stopped now 2 years into treatment. This is a huge relief as these episodes were scary. Also, I am able to eat quicker than I did before which means I'm finishing meals when they are still hot.' Adult living with type 3 SMA

'I used to aspirate my own spit at least 3-4 times a day, this used to lead to debilitating coughing fits lasting between 30-45 minutes each time. Now this happens to me once a month if that. Also, eating was becoming scary and I had to think carefully about what I was eating so I didn't choke, I no longer have that worry. 'Adult living with SMA type 3

Fatigue

More stamina throughout the day. This is making my working day so much easier! I used to want to cry in pain from a day at work, typing on laptop - not anymore. I no longer feel that 'withdrawing from work' is on the horizon and I can earn a good income for at least another 10-15 years provided the [treatment] benefits are maintained. Adult living with type 2 SMA

Before treatment we loved swimming but the fatigued was intense afterwards and she started school and was super tired. Treatment has helped her tiredness levels immensely. She is such a happier child. By bedtime she use to be asleep quite quickly from being so fatigued but now we have a bedtime talk and read a book - which is aiding her education' Co child living with type 2 SMA

‘After a full day at school he is definitely ready for a lay down and rest. Which is better than he was where he wouldn't be able to do full days due to tiredness.’ Co child living with type 2 SMA

Mental Health

You need to understand little bit about what life growing up with SMA is like, when *you are young you are told you will die in your teens… maybe… if you're lucky (or you were told this in my day), then you get through those years and you're told you might get to 30 odd… Then you get to that age and you start thinking "when am I going to die".*

When you're young you don't care, you live life like every day is your last and it's not so bad. As you get older, in my thirties I was living with my partner and having children, I had responsibilities and other people to think about. This really messed with my head.

Then I had another consultation with a different doctor and they told me that I was probably going to live until I was 50 or so, this really changed my thirties, but seriously impacted my forties and even though I was working full-time senior management role in a government organisation and was in charge of millions of pounds and lots of people working for me, I couldn't stop thinking I was getting close to dying.

Then this drug comes along, I couldn't get it then but something changed, there was hope. There had never… Ever… Been that before for people with SMA. It was hope for my wife, was hope for my children, hope for my friends and other relatives who, in some regard, all expected me to die… This seeps into your consciousness, you start to expect it yourself, and now maybe not… It's weird thing to suddenly have a future.

And then it was YEARS before I could get my hands on the drug, my hands started to go, I couldn't drive my car any more, I couldn't even drive my chair reliably any more. Breathing was becoming difficult sometimes, choking was happening more often. I felt like I was slipping away without being able to do anything about it… Then I finally get access to Evrysdi and within 2 years, even though I'm much older, I feel so much better. I have stamina, I have strength I had lost, I can breathe, I don't choke, I can eat and drink without worrying about killing myself… Can you understand this? Can you understand I had to think about not dying every time I sit down for a meal? I hope you can't, but you need to think about that fact, that silly singular little fact.

Now I can also drive my chair again, that most fundamental of things, the ability to move yourself about at will, that was restored to me. That is such an important freedom to have received back when it was almost gone.

It hasn't just positively impacted my mental health, it has, and I mean this quite literally, given me back my life. Given my family a major breadwinner, a husband and a father back to them, it's allowed me to believe I now have a future, and that I will also see what the future holds for my loved ones as well.

I don't know if there is a big enough word to describe what that means?

’ Adult living with type 2 SMA

Quality of Life

There is hope and there is evidence via the assessments that I am not deteriorating. I can cut up my own food now, eat more than soft food only, brush my hair now, I feel like a person again and not simply a burden. I have children and want to be there for them, this ***drug gives us all the life*** *we want. Before Spinraza I was losing strength and stamina and my children were missing out in comparison to other children.'* Adult living with type 2 SMA

'Loss of strength and progression of my condition is no longer a daily worry. Access to a treatment, that is clearly making a positive impact on my physical health, has given me hope and the freedom to live my life without constant concerns around the future.' Adult living with type 2 SMA

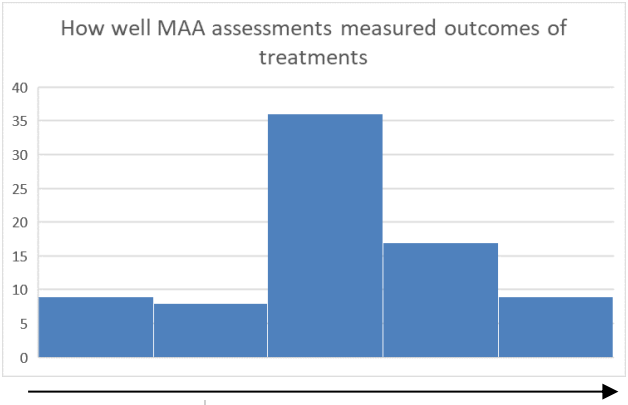
'I can plan for my future, thinking about the next 5 or 10 years without wondering if I'll be significantly weaker by then. I'm able to make decisions about my life based on how I feel now, instead of how I'll feel if things get worse.' 20-30 yrs type 2 SMA

5. MAA tests and assessments

How well do you think these tests and assessments worked in measuring the effectiveness of these treatments?

5:1

Living with SMA:

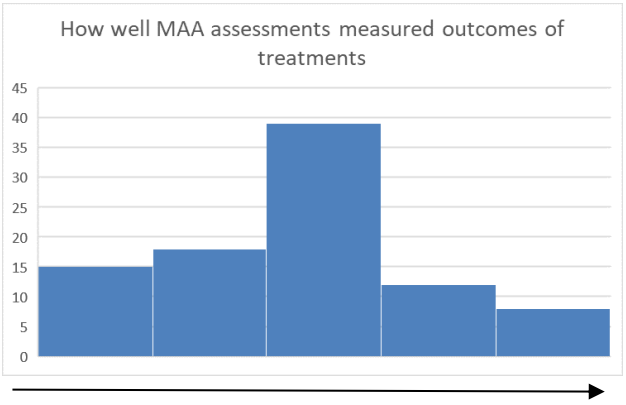


They **did not reflect** how the treatment has impacted daily life.

They **somewhat reflected** how the treatment has impacted daily life.

They **totally reflected** how treatment has impacted daily life.

Carers:



They **did not reflect** how the treatment has impacted daily life.

They **somewhat reflected** how the treatment has impacted daily life.

They **totally reflected** how treatment has impacted daily life.

5:2

'They [assessments] are a box ticking exercise. The small, but significant improvements of someone with type II in later life (over 40) are almost immeasurable, yet represent a positive, life changing benefits.' Adult living with type 2 SMA

The tests are weather dependant- my hands don't work as well when its cold -The location of tests was different each time so I performed differently. For example, on my last test I couldn't reach the table very well so didn't perhaps perform as well as the time before, when I could access the tests better - The test where you have to open the Tupperware pot had a different pot to open one of the sessions meaning the tests weren't fair' Adult living with type 2 SMA

'The examinations seem to test me on such random abilities that I will never and have never used. The things that are important to me our typing speed, swallowing, coughing, swimming and maintaining neck strength. How far I can extend my leg when I'm sitting in my wheelchair has no impact on my daily life.' 15-20yr old living with SMA type 2

6

Case Studies

6:1 Spinraza

Arthur

Arthur was diagnosed with SMA type 2 at 18 months old, we had concerns at around 9 months but were told not to worry until 12 months. At 12 months we made an appointment & had a 6 month waiting list to be seen, a few days before the appointment Arthur was admitted to hospital with a virus and the doctors were very concerned about his lack of mobility and floppiness. Shortly after we received the diagnosis.

At that time it was absolutely devastating, reading about SMA we were absolutely broken. We had no idea if we could keep working, if we would lose our house and how were we going to cope watching our son deteriorate before our eyes.

SMA has had a profound effect on our lives as a family. We have to juggle lots of appointments, for check ups, treatment, Physiotherapy, hydrotherapy & Hippotherapy.

We have faced issues with Arthur's nursery & him not being able to be with his peer group due to the class being upstairs and health and safety issues with no 121 support being offered by the council, so he has to stay in the baby class until we can get him into pre-school. This is detrimental to his cognitive and communicational skills as SMA does not affect his intelligence, he has struggled seeing his peers move up to the next class.

Financially it is extremely tough, we have spent a lot of money adapting our house and garden for Arthurs needs. We have also invested in equipment both at home and nursery to aid his progression. We also had to buy a bigger car for his equipment as he is under 3 years old he doesn't qualify for Motability scheme.

Living with a child with SMA is emotionally draining, I have recently contacted the doctor for counselling and treatment for stress and anxiety. It is a constant worry about the future and his independence.

Treatment makes this bearable, of course this isn't the life we would have chosen for Arthur, however the access to treatment has been lifechanging and gives us hope. Arthur initially lost strength before his diagnosis and has gone from only being able to roll & sit without being able to reach out for anything to crawling, standing & walking in his walker. Who knows what the future holds if the drug is approved as he is going from strength to strength. I believe he will be able to walk and gain more independence to do things for himself if the drug is approved.

It is inhumane if these drugs are not approved after the clear benefits this brings. Arthur is gaining strength every day and without this I don't know how I would be able to watch him deteriorate; it just doesn't bear thinking about.

Maxwell

Living with Spinal Muscular Atrophy is challenging at times, but the person and mindset created by it makes it a little easier. When thinking about what it's like to live with SMA, it's important to think about emotional health, created by our actions.

I'm currently using a £20,000 power wheelchair that was funded privately due to the NHS's solution being adequate. In fact, much of the equipment I used to live with was funded privately or donated. Including the gym equipment, I use in pair with my Spinraza to slow the progress of my condition. Often, I don't think about my overall strength suffering from SMA, but treatment has given me hope to try and not lose muscle mass. I am weaker being type two SMA, which means I need others to fulfil physical tasks for me, like dressing myself.

I currently rely on twenty-four-hour care, which is shared between the carers I employ and my family. Independence is a factor that contributes the most to health and it can be hard in a care crisis to have your needs and wants fulfilled. I currently have one carer who took a few weeks to train in hoisting, dressing, hygiene, cough assistance and much more. When my carer isn't around, I rely on my mum who has recently gone back to work after I fought for a big enough social care budget to be independent in the day.

I currently go to university and am a manager, entrepreneur, and social media influencer. I'm proud of all the things I do, but I am a manager not out of choice but because I must. In managing my own care team, wages, worries and paperwork. I've been in a wheelchair since the age of two years old, and my parent, not long after, embarked on a 5-year battle to get my house adapted, which they later ended up paying over £ 100,000 for. This life-changing renovation allowed a full wet room with hoists, changing tables, adjustable sinks, an adapted bedroom with a through-the-floor lift and an adapted toilet downstairs.

To get to the point I am today has been a journey of breaking social norms and fighting a system which is not fit for purpose. However, in many aspects, it made me the person I am

today. I’m an A* student with a 60% attendance due to illness and hospital appointments. Which I never let get in the way of my learning and sparked research and learning at home. This led to me becoming very entrepreneurial, sparking a love for being curious and exploring different avenues. Now I volunteer for three charities, am part of a future board changing my local town, I’m a leader of a national campaign, am a business management student and have run my own business.

I have a strong friend’s group who are extremely supportive. When we want to do something together, we do it. My closest friend spent weeks planning an accessible night out in Sheffield for the group. While I also cater to their needs, showing a reciprocal bond of true friendship. They are strong advocates for disability and social change, just like my partner. Relationships is a topic not talked about by the disabled community often. It’s a roller coaster navigating a space in which there is no information widely spoken about, but we are extremely close, being together for five years. It's something that’s truly very important to me, a healthy relationship and something I'm proud of. Although I’m smart, debatably good-looking and determined, society has told me I can't be those things all my life. So, sometimes, it doesn’t really feel real.

Summarizing the experience of living with SMA in a single-page document is daunting; I would need a few books; it's a narrative that spans volumes. However, amidst the challenges and stereotypes imposed by society, the triumphs and joys of overcoming or debunking them shine brightly. Bright enough for me to be more determined and inspired than my peers, to build my dream life and, therefore, experience.

When first receiving Spinraza me and my family were extremely happy. To finally have a medication that would slow my condition that appeared to come out of nowhere. It didn’t disappoint, either. When first taking it, I instantly felt stronger; I didn’t know if this was a placebo effect or not. My voice appeared to get deeper, and I felt energised and fatigued slower.

It was not a placebo; in my first lung function test after treatment, I had increased my lung capacity by a few per cent. I then beat the strength test by a point in the following months. My strength is consistent two years later, and I often feel ready for my next workout. From a test-scoring view to me and, my family it's easy to see the work treatment has done for me over the last two years.

Although the test is just one part, it is not so important to me. At first, I didn’t realize my strength; mum would say, “maxwell, you just moved that plate”, for example. Suddenly, without thought, I would be doing harder tasks than I normally do. This later became evident to me when I found it easier to put on my gaming headset. This rush of energy sent me into a fitness era where I would innovate and adapt routines for myself to try and build strength. This then positively impacted my mental health, I felt like I was on top of the world.

Now, all I wish was I could get more! I feel dips in between treatments. For me the only inconvenience is travelling to London for the treatment. Which is a small price to pay for the motivation strength and hope Spinraza has given me. I know many case studies are different, but evidently without the treatment I would have declined by now, not improved. This is all I wanted, this is the end goal for me.

Miss P

Miss P was diagnosed with SMA (three SMN2 copies) in February 2018 whilst we were on an international work assignment living in America. She was 2.5 years old at the time, began nusinersen treatment immediately, and remains on treatment today under the MAA.

Although there were significant signs of SMA early on, nusinersen arrested her first major decline. Prior to treatment Miss P was tired and fatigued, missing pre-school due to regular coughs and colds of increasing severity, unable to bend her knees whatsoever, and finding getting up from chairs, on and off toys, stairs, and uneven surfaces more and more difficult. Life for all of us was a mire of endless medical appointments, assessments, and trips to the pharmacy. As her only carers my wife and I were distraught at watching our beautiful daughter struggle and literally fade away in front of us.

Due to treatment Miss P has had an HFMSE score increase of ~30 points.

Today, despite normal growth and development (Miss P has gained 20 kgs and 40 cm since diagnosis), Miss P is a happy and active little girl who fully participates in every opportunity at school, rides her bike, goes horse riding once a week, and to a Stage 7 swim class on Wednesdays. Miss P has a bright outlook on life, is rarely affected by circulating coughs and colds and we attend hospital a handful of times per year for nusinersen doses and MAA assessments.

Some things like running and jumping still look different but with ongoing treatment we are hopeful that the effects of her SMA will not hold her back and will enable us all to live a near normal and happy family life.

Should treatment not be approved in England Miss P would deteriorate as this savage disease destroyed her physical ability and quality of life. Watching your child decline in this way knowing it is entirely avoidable is an immense cause of distress and anxiety. If nusinersen was not available the wellbeing implications for my wife and I would be severe, our lives would fundamentally change in terms of capacity to work professionally (and pay taxes), we would need to sell our home to move to amenable accommodation, Miss P would need to move school to one without stairs, and we would lose the simple enjoyment of every day family life that most take for granted.

Sinead

My life, like most others, has been shaped by numbers. Onset of condition at 2, initial diagnosis at 8, broken pelvis at 25, confirmation of SMA type 3 at 30, broken arm at 33, number of falls over lifetime – thousands, near misses – innumerable , first Spinraza treatment at 46, reduction in falls 95%.

But numbers only tell half the story, and sometimes even less. They don't capture the years of worry for my parents, who knew something was wrong with me but who nobody would listen to. They don't show the little girl at primary school, falling again and again and again -

painfully learning what it felt like to be the odd one out. They don't represent me at secondary school – tying to fit in while falling down, not being able do the same things as my peers, having to leave lessons early to try and pull myself up the stairs to the next classroom. Numbers can't come close to showing how I battled through university – exhausted, bruised, often bleeding – determined to keep up, to qualify as a Speech and Language Therapist. They can never capture the arc of my life as a person with SMA - as a partner, a daughter, a clinician, a colleague, a patient. Numbers will never, ever be able to delineate the way Spinraza has changed my life.

To live with SMA without treatment is to be engaged in an almost constant negotiation with your own body, trying to balance the equation of fatigue, ability, pain, achievement, fear, frustration and risk in order to do the most benign activities – going to work, cleaning the house, getting out of bed. For me, SMA is the ever-present background hum of fatigue and pain, interpolated with sudden, unpredictable falls, where my legs give way with no warning and I have no ability to save myself. Along with these are the falls caused by slight changes in terrain, trips, slips and stumbles that others would recover from without even noticing, but which I cannot mitigate for and come crashing down. Falls can and do happen anywhere - crossing a busy road, in the supermarket, in the middle of a clinical session with a patient. At best the results can embarrassing as I crawl on the dirty floor looking for somewhere I can get myself back up, wondering if I *can* get back up. At worst, hospitalisation.

As the subject matter expert on my own condition, I have learnt the hard way how to best manage my life to make the most of what I can do while minimising, but not excluding risk. Every day, every hour I am dynamically assessing what I can do, testing the limits of my body, making deals with it – if I'm on the wards today I'll be tired so can't go swimming, tomorrow I'll go to bed at 7 so I can drive to visit my parents. SMA is costly and demanding and is getting more so. From my 30's I was aware that I was slowly deteriorating as ageing and sarcopenia took its toll on a body with a muscle wasting disease. The cognitive load of SMA can be exhausting, let alone the physical effort required to live, and get the equation wrong and the consequences can be devastating.

As such, while the quantitative data collected as part of the MAA provided a level of evidence and insight, it can never capture the complex, nuanced and subjective experience of having SMA and receiving treatment for SMA. The reliance on motor function assessment such as the HFMSE and the ULM for such a diverse patient cohort, dampens the potential data impact. They are repetitive, blunt tools which lack the flexibility or dynamism to capture meaningful, functional subjectively and objectively experienced change. Repeating them en masse and in person every 6 months is a significant and frustrating burden to patients which may act as a barrier to engagement.

What that data collection fails to capture is the global impact of treatment on the patient. This is not just a 'slicing up' of motor functions, or distanced walked, or coins picked up. Rather it is something greater than the sum of its parts - a life lived better and longer, with less fear, less risk, less pain. It is as if the scale of 1 to 10 pre-treatment becomes 1 to 20 post treatment as life and health and ability and possibilities expand exponentially and

unknowably – how can you possibly know all you can do now, any more than you could know all you couldn't before?

Since receiving Spinraza, the horizons of my life have shifted. I have stopped falling without a cause and hardly ever trip. I have not broken any bones, my knees are not constantly scabbed and bleeding, I don't have to explain away bruises. I can walk further and faster and for the first time in my life I can walk with a pleasure, literally enjoying the view. I can engage in more strenuous exercise and reap the benefits thereof. I am stronger, more stable. I can work harder and for longer and see more patients. I'm more independent. The deterioration I observed with a terrible knowing stopped and in fact I suddenly felt better in a way I could not have dreamt of. Since receiving Spinraza I am less afraid of the future. I don't have to keep my eyes down, watching for each step I take. I don't have to hold on to my partner to stabilise, I am literally less of a burden. I don't have to dread the terrible feeling of falling with no safety net. I'm not waiting for the fall that will tip the balance and cause my world to negatively pivot. My life is easier and safer and I am happier and more productive. I am me.

But back to the quantifiable – if I construe an alternate reality where I am not receiving Spinraza, what does that look like? My deterioration has continued. It is highly unlikely that I will be able to work at my current level or work place, providing highly specialised speech and language input for 40 patients with traumatic brain injury, supervising junior clinical staff and teaching. My partner will have to take on a significantly increased caring role, which would impact on both her work researching positive ageing outcomes, and the nature of our relationship. I in turn will no longer be able to maintain caring role I play for my parents, forcing them to rely much more on statutory services. I will lose my current ability to exercise and so will experience the range of negative health outcomes associated with increased inactivity. My mental health and that of my partner is likely to suffer. As I age without treatment, the frequency and severity of my falls will increase rapidly, leading to much greater use of primary and a secondary care services. I will attend A&E much more frequently, often via ambulance, and will require surgical interventions, rehabilitation services and equipment. After each fall I will never quite return to the level of function I was at. My home will either need adaptations or I will have to move to specialist housing. Soon I will require an ongoing care package. One day one of my falls will likely kill me.

There is also another version of reality at play, one in which the little girl didn't start falling down, where Spinraza was available in 1975. SMA has framed every aspect of my life, from the jobs I have taken to my decision not to have children. It has shaped my relationships, my thoughts and dreams and not a single hour has gone by when I have not been aware of it. If I could give that little girl the gift of a different life would I? If I could take away the pain, the broken bones, the bruises, the humiliation, the fear would I? That little girl has made me who I am today, and I am proud of that, but I'd spare her in heartbeat.

It feels almost impossible for me to convey to somebody who hasn't experienced it what it means to have your life changed by treatment. Imagine you wake up one day and can fly, after years of dreaming and wishing that you could. Try putting that in words. Try having that judged and quantified. Try having that taken away.

Steven

My name is Steven Jones and I have SMA type 3. I use a wheelchair full time, but live a majority independent life in terms of living on my own, driving, going to work and I currently work in the civil service.

I started Spinraza in the summer of 2022, and this was a really interesting time for me, I've never really seen my disability as something that needs treatment or needs support. But throughout my twenties, and I'm 30 years old now, I had seen not s rapid decline but a very noticeable decline in my abilities. In my early twenties I used to be able to get up, go around the house on my knees if I wanted to, go upstairs, I can't do that now . I also started to find transferring really difficult and really noticed when the height of the bed, chair or toilet was a bit lower, it was really difficult to get back into my wheelchair. That caused me quite a lot of stress and anxiety thinking, if I'm going to public places, what would I be able to do?

Getting treatment was quite a difficult decision for me because I was thinking, do I need it? What will be the side effects of having a lumbar puncture? I went for Spinraza because that provided more certainty because of the data at the time compared to Risdiplam.

So leading up to treatment, we were just coming out of lockdown which I found quite difficult for my body because instead of going to work every day I was sat at home and I think with SMA, if you don't do certain activities you lose the ability to do them. I was really tired every day, I found it really difficult to get out of bed, I didn't really know what my fatigue level would be from day to day, what I wanted to do or what I could do. That provided a lot of uncertainty for me.

I used to avoid going out because I used to be worried about the accessible bathrooms, would things be the right heights? Will I feel really tired? How will that impact on my day?

Once I started Spinraza, things started to look a bit more hopeful, I had a noticeable increase in energy and fatigue levels dropped. Of course that wasn't instant, but looking back compared to what I was like at the end of 2021 start of of 2022, I now have a lot more energy and I'm able to continue to live an independent, fun and happy life. That's been the biggest noticeable change. I haven't noticed the impact of transferring anymore so that would be another noticeable change.

And secondly, a bit of hope, those in the SMA community have to come to terms with change in their disability over their lifetime. I'm expecting, as I get older, my abilities to decease even further and that's quite frightening. I think; What will I be like when I'm 40 years old? 50 years old? Will I be able to work when I need full time care? But what Spinraza does, it helps to stabilise the condition and I've noticed that stabilisation in my condition. That has enabled me to hold a bit of hope for the future, hope that I will be able to live a full life, go on lots of holidays and continue to work, continue my social life and my hobbies and activities. That is invaluable and I think that stabilisation is really the hidden benefit of this medication. Its not about helping me to walk again like I used to in my teenage years, but actually that stabilisation is so valuable to me as it keeps me independent which is actually such a full part of my identity and who I am.

Looking back, I would not have changed my decision to get Spinraza, I find the lumbar puncturescompletley fine, a couple of minor side effects but they are not really noticeable, I can almost forget about the medication for four months, go back to work or on a holiday, enjoy my life knowing it is coming up in 4 months and have a bit more certainty for the future.

Video version - WIN_20240224_09_33_59_Pro.mp4

6:2 Risdiplam

Anonymous

You need to understand little bit about what life growing up with SMA is like, when you are young you are told you will die in your teens… maybe… if you're lucky (or you were told this in my day), then you get through those years and you're told you might get to 30 odd… Then you get to that age and you start thinking "when am I going to die".

When you're young you don't care, you live life like every day is your last and it's not so bad. As you get older, in my thirties I was living with my partner and having children, I had responsibilities and other people to think about. This really messed with my head.

Then I had another consultation with a different doctor and they told me that I was probably going to live until I was 50 or so, this really changed my thirties, but seriously impacted my forties and even though I was working full-time senior management role in a government organisation and was in charge of millions of pounds and lots of people working for me, I couldn't stop thinking I was getting close to dying.

Then this drug comes along, I couldn't get it then but something changed, there was hope. There had never… Ever… Been that before for people with SMA. It was hope for my wife, was hope for my children, hope for my friends and other relatives who, in some regard, all expected me to die… This seeps into your consciousness, you start to expect it yourself, and now maybe not… It's weird thing to suddenly have a future.

And then it was YEARS before I could get my hands on the drug, my hands started to go, I couldn't drive my car any more, I couldn't even drive my chair reliably any more. Breathing was becoming difficult sometimes, choking was happening more often. I felt like I was slipping away without being able to do anything about it… Then I finally get access to Evrysdi and within 2 years, even though I'm much older, I feel so much better. I have stamina, I have strength I had lost, I can breathe, I don't choke, I can eat and drink without worrying about killing myself… Can you understand this? Can you understand I had to think about not dying every time I sit down for a meal? I hope you can't, but you need to think about that fact, that silly singular little fact.

Now I can also drive my chair again, that most fundamental of things, the ability to move yourself about at will, that was restored to me. That is such an important freedom to have received back when it was almost gone.

It hasn't just positively impacted my mental health, it has, and I mean this quite literally, given me back my life. Given my family a major breadwinner, a husband and a father back to them, it's allowed me to believe I now have a future, and that I will also see what the future holds for my loved ones as well.

I don't know if there is a big enough word to describe what that means?

Jay

My son Jay Pickston, will turn 16 in March.

As we are all aware SMA is a completely muscular disability. My son is extremely intelligent and he is on course to hit all his GCSE targets. He has already been accepted at a 6th form college, where he will study A-level Law, English lit and Criminology.

So the need to be as fit as possible and as independent as possible is so important to all people. Biased or not, especially in the cases of physically disabled people.

He is clever and I even think the teachers at his main stream school where amazed at how intelligent he is. Maybe the electric chair deceived them? We still live in a judgemental world. Although I'm proud to say in my small world this is decreasing.

As well as being clever – he is street wise. He has a great group of mates that are all popular within the high school and around the town. I never worry that anyone is going to pick on him, bully him etc.

Jay gets on the bus on his own and with friends to go to the soccer factory in Rochdale, into Bury for food, shopping, cinema. Down to the local sports centre to watch his mates kick a ball about and play badminton. Even as far as the Trafford Cnt.

He really is strong minded and independent.

So Risdiplam – OMG – where do I start. To take his drug away and then I could only guess would reverse all the good that has come out of it and see my little man deteriorate, would like being diagnosed all over again.

As I have said, Jay will always try to do things himself. Always. If he says he cant do something, then he 100% cant do it. We have the 6 monthly physio assessments and these have improved everytime. To the lay person, these improvements might seem small. Maybe insignificant.To those who know the SMA world, they are huge.

So I don't have to hand or know the exact measurements from the official physio sessions at RMCH. But our findings from home are:
He could only move his index finger and thumb on left hand. Now he can get this hand straight and wiggle all fingers. His right hand he has full range. So in the winter now, he can put his own gloves on. Even I used to struggle when he was younger and all through out life as his hands didn't go straight.
He is addicted, and also very good on his PS5. As long as I keep the wire in reach, Jay can plug this in when it starts to die.
He can bend over in his chair and untie a show lace. Yes not taking the shoe off, but can pull the lace to untie. And then sit back up.
If we are not going out that day, he tends to wear football shorts. If in these, using his uri bag, he can give himself a wee.
We have an adapted bathroom, but he can now turn the taps on and off to wash his hands.
If the carton/bottle isn't full he can pour himself a drink into a glass.
Jay still chooses to hand write in lessons not needing timed exams etc – and his hand writing has massively improved.

I don't know – there is probably a whole lot more that even I now take for granted. And although there are still limits eg the drink carton/bottle not being full, having to be in football shorts. These are massive to my son, myself and our world.

This drug is a wonder drug. And doesn’t involve infusion. I can only pray the funding continues. I have just signed the consent form so that all data can be shared now world wide, rather than just UK to have a larger research field. Fully support this.

Come on world! Any improvement can not be given then taken away.

Hanna

I have been taking Risdiplam since April 2023. During this period my muscle strength and physical capacity have increased drastically. I feel way more energy than I used to. I used to feel strong fatigue almost every day in particular after I spent a day out. But now I can be out for the whole day and feel okay the next day. It allows me to study at the University full-time as well as have a placement at the gallery. It is easier for me now to travel too. This summer I was on four-day trip to Liverpool with very action-packed days. Without the medication, I would have felt terrible and probably wouldn't been able to go on such trips. But with Risdiplam I felt just a bit tired when returned home. It means the world to me because I would like to work at the gallery soon and now I feel like I can do it.

All my friends and relatives have been noticing that my voice is becoming louder and louder. Now I can speak for a long time without being out of breath. It means a lot to me because I was concerned about my lungs' health before.

But even in the case all of these improvements hadn't happened it would mean a lot to me it went to keep my health condition stable. Because I had been observing my strength decreasing. And I'm sure that without the medicine I will lose the ability to paint in the next five years as I am an artist.

Taking the treatment for SMA is essential for my mental health, future work, life quality of all members of my family, and my freedom. Because I am going to be the working member of the family and support my parents.

Anonymous

I have been taking Risdiplam since the beginning of the EAMS in December 2020. There are two areas in particular that I would like to highlight in this case study. Firstly, the physical element of the treatment.

I work full time and at the end of 2020, following what had been an extremely tough year with the pandemic, I had deteriorated to a point where I felt it was almost impossible for me to go back to work. I had little energy, little strength, I was struggling to drive my wheelchair and got extremely tired and suffered from extreme fatigue even just working for short periods. I had all but given up all hope of returning back to what had been a successful career. However, I was given the opportunity to receive the treatment and I can honestly say it was life changing.

Within a few months I had regained my energy, some of my peripheral dexterity had returned to the point where it had been earlier in the previous year. I found my difficulties with fatigue

had almost entirely disappeared and I was now able to get back to work. I felt confident and empowered to continue my job and have even been able to secure a more senior role since.

In terms of the emotional/mental health aspect, the benefits are immeasurable. While I acknowledge the treatment is unlikely to provide me with significant improvements in my condition, the fact that I can now look forward to stability and maintaining my current levels of energy and strength has had unbelievable impact on my mental health. No longer am I fearful of which function I am going to lose next, no longer do I need to worry about choking every time I eat, no longer do I need to worry about getting stranded somewhere because I'm too tired to drive my wheelchair, I could go on.

The impact the treatment has had on my life, and those around me, cannot be summarised in words. SMA is an evil condition, one that condemns you to lifelong grief and sadness due to your constant decline. Finally, we have treatments available that can give me back what is left of my life.

Lucy

My son George aged 12 was diagnosed just after his second birthday with Spinal Muscular Atrophy Type 2. George has never walked and only stood with the help of orthortics and a standing frame.

In George's early years he was a very poorly little boy often ill with chest infections. He was underweight and very frail. I lived in fear of George getting ill and often avoided social settings including days out, gatherings, parties and even shopping. Unfortunately this was mirrored in his sisters early childhood who was born only eleven months earlier and we all lived a very isolated life.

His time in a educational setting was greatly reduced due to his own illnesses and also avoiding illness should it be in the classroom. His average attendance was around 27%.

George was gradually losing physical strength. He lost the ability to crawl, get on all fours and play with many of his toys in the first six years of his life.

George's respiratory became weak with his lung capacity dramatically falling every six months at his reviews. At the age of six a videofluroscopy showed George was aspirating on his fluids.

SMA took its toll on George and also the family effecting our mental health, finances and pressure that was felt with the need to find suitable accommodation, equipment, wheelchairs etc. Attending appointments which only vocalise the decline of your child was extremely hard. SMA is not a static condition so living with the constant fear of lose for your child was extremely hard.

After George started taking Risdiplam everything changed. Everything.

The first thing (and for me the most important) was that George recovered his swallow and his lung capacity doubled. This is not only life changing but life saving.

It meant less illness, weight gain, more time in school, a better social life, less worry - equalling a better mental state. The fact that my child would not need permanent NIV (or even just night time NIV), nor would he need to have a peg fitted was a game changer forever.

Also the real fear of death. The fact that a cough or even a cold could have killed him. Of course I will always worry when he ill and he is more vunerable then most kids his age but the real risk of death wasn't there anymore and that is priceless.

George was physically stronger. He not only stabilised but he gained. Opening packets of food, washing himself, putting his own shoes and coat on, cutting his own food....the list is endless. To simplify it George just became more independent and that independence has had a knock on effect for me meaning I do less and his sister now gets more time with her mum.

As it stands now George attends a mainstream senior school and has a attendance of one 100%. He is one of the top students in his year and excels. Had his attendance sat around 27% this would not be the case.

George plays the drums and is doing his grade one this year (bare in mind prior to Risdiplam he could not push lego pieces together). He is also doing a public speaking exam, his projection and diction has greatly improved with the drug.

George goes fishing and can unbelievably cast a fishing rod out and he enjoys drawing (which he struggled with before).

Since starting Risdiplam George has not been admitted once into hospital compared to the countless times before. Risdiplam saved my sons life and changed it for the better. It also changed mine.

Mark

I have been taking Risdiplam for nearly 2 years now, and that I feel it has made a significant difference. Before having Risdiplam I was losing strength quite rapidly; I had concerns about my ability to continue to use a mouse - left clicking was getting more difficult, concerns for swallowing, and deterioration in my breathing.

Risdiplam has stabilised my condition. It has also given me small improvements. For instance, I lost the ability to right click my mouse a couple of years ago, but now I am able to do this again.

Rather than continued deterioration, Risdiplam has given me a future.