

Appendix 7.

Survey Jan – Feb 2018

Children treated with nusinersen – impact and expectations

1. What impact has the treatment had on your child?

Type 1, treatment started < 7 months, 5-7 injections

Nusinersen has had a dramatic effect on the quality of life my son has. Before treatment he could not even grasp - now he can use both hands to play with toys, passing from hand to hand he is beginning to hold his head up and can move his legs a little. He has been managing colds all through winter at home whereas before he was in intensive care on life support for every cold he got. He is a happy boy who can now start to explore his surroundings, he is also beginning to talk and can say Mum and dad and can sing and clap

Type 1, treatment started age 8 - 12 months, 5-7 injections

A strong impact, in the good way. She shows signs of improvement in her motor skills, she is a happy child because she can move her hands to play around. She is happy because she can follow me by moving her head from side to side...

Type1, treatment started age 13 - 24 months, 5-7 injections

Physically she has gained strength, particularly in her head control and core strength. Emotionally she has been very resilient. Her understanding is limited by her age, but treatment has caused her minimal upset. She has coped extremely well. Practically she is able to perform more tasks herself and gained strength to use her own wheelchair.

Type 1 treatment started 13-24 months, 5-7 injections

My child required/relied on bipap before treatment and her lungs were getting worse and worse. We as parents had made the hard decision to not trachea our child for her quality of life and were preparing ourselves to lose her, as she constantly caught virus/infections and spent most of her 1st year of life in hospital. However, nusinersen has stabilised / improved her breathing. She now only requires bipap for sleep and her settings have been turned down following sleep studies. She has gained some strength in muscle tone throughout her body, mainly her trunk/arms/shoulders. Her neck is now also showing signs of regaining strength although not able to hold her own head yet, she is beginning to and is turning head herself. Legs/hips have also v slight signs of muscle movement. But most importantly overall my child, is much more content no longer working so hard to breathe and she is a much happier / content child with smiles and laughter.

Type 1 treatment started age 8 - 12 months, 5 - 7 injections

The treatment has shown physical improvements for our daughter. She has not shown any progression of the disease since treatment started and has gained more movement allowing her to play with toys. This is a big positive impact on the quality of her life....

Type 1, treatment started 8 - 12 months, 5 - 7 injections

My daughter has gained wrist and lower arm movement she now has a wonderful strong hand grip she tries to lift can't gravitational lift but she now tries. she has ankle and foot movement. She moves her shoulders and attempts to lift her head amazing to see she lost all movement. Vocally she has got very loud can say dad, es(yes) and she babbles which is really trying to talk. Hasn't helped majorly respiratory but we are working on that.....she has tolerated every treatment amazingly and she fights day in day out wanting to try more and more tasks with a smile on her face

Type 1, treatment started <7months, 11+ injections

Physically he has gained so much strength in comparison to how he was when he started. It's been slow and steady. He can now move his legs, grip better, lift his arms against gravity, assisted roll, play with toys, Hold his head with minimal support. He now has a power chair which he controls himself. He can tolerate sitting up for hours without any respiratory support. I can cuddle him! Respiratory wise he has gone from being ventilated 22 hours a day to 16 hours a day. Emotionally he has always been happy and content but obviously now with his new-found movement and abilities he is very proud of himself. He can tolerate doing things non- affected children can do like going on rides and playing. This makes for an even happier boy! Practically he can do so much more. SMA has never held him back but now he can do so much more making everyday life much more practical.

Type 1 / 2 treatment started age 13 - 24 months, 0-4 injections

She has gained skills whereas before treatment she was just losing skills. She has gained head control, more movement in arms and legs. She is able to roll forward which was something she could never do. It has given us all hope. She has stayed off respiratory support and feeding support

Type 1 / 2, treatment started 37 months +, 5-7 injections

After years of deterioration and hearing that everything is getting worse at every review, this year for the first time our daughter heard that she's doing better, both at spirometry and CHOP. This gave her hope that her life can improve, the trouble of stretching and physio is worth it, and there is a future for her. Her biggest joy is being able to cough better, and deal with mucus plugs without so much chest physio and cough assist. Also, previously every illness (respiratory or gastric) meant non-reversible deterioration, and now she bounces back almost to the same level as before the illness.

Type 1 / 2 treatment started age 37months +, 5 - 7 injections

She has gained 5 points on her chop test! We have seen improvements in her neck/head control, the trunk of her body and she can do a few more movements with her thighs, she can also lean without flopping. Emotionally she's doesn't like the procedure but our hospital have been great offering extra support

Type 2, treatment started age 4, 0 - 4 injections

Strength, gradual improvement in motor function, general health improvement

Type 3, treatment started age 12 years, 5-7 injections

We have seen significant improvements in his health with Spinraza treatment. Physically: He doesn't fall/collapse as he did before treatment. He fell at least twice a day & some days multiple times. He can now walk faster/further, his gait has improved & is less waddling. In the 6-minute walk test at GOSH he walked an EXTRA 32metres in December2017 than he did in June2016. During this time, he had grown in height & increased in weight which, without treatment, would negatively affect his walking ability. He has improved in other motor functions, he's stronger/has more stamina/doesn't fatigue as he did before Spinraza. He can cycle on the exercise bike and getting better/faster with every treatment. Before treatment he had begun degenerating to the point where if he fell in the middle of the room he would crawl/drag himself over to a chair to assist himself up. He can now independently rise from the floor again & with each treatment he is becoming obviously better at this & stronger. Before Spinraza treatment he had become reliant on a carer to put on socks/shoes/splints. He can now do this himself. Spinraza is increasing his physical ability to be independent. He had bilateral pronated flat feet before treatment—he had pressure areas & pain. His right foot has developed an arch & no more pressure areas/pain. This has also enhanced his walking ability. Emotionally: SMA has ceased to progress since he started Spinraza. He is becoming increasingly able and independent which is positively affecting his attitude to life. Before treatment he regularly fell/collapsed causing him severe pain. He had bad falls where he couldn't weight-bear requiring A&E treatment: He had Metatarsal fracture & Soft tissue Injuries over the years. Each bad fall resulted in his being petrified that he would NEVER walk again. He is NO LONGER scared of losing ability/getting weaker. He is embracing life and is now developing without fear, he's stronger, more stamina & MORE ABLE as he grows. He has a thirst for knowledge & life. He is exceptional in all subjects at school. He wants to study Law and become a Lawyer.... Practically: Before Spinraza treatment he had become increasingly reliant on his power wheelchair to get around. With treatment he uses his wheelchair less over time....

Bereaved: Type 1, treatment started age < 7 months, 0-4 injections

Physically: It did not affect him significantly giving him the injection. He did not reach any milestones. He developed the ability to move his forearms for short periods against gravity and in his legs small movements, but not against gravity. We have since learnt that this may not have been a reflection of the benefits of Nusinersen as our Neurologist explained that babies become relatively stronger as they grow. Emotionally: Caused him to cry for a matter of minutes to administer the drug, it had no emotional impact apart from this, he was characteristically a happy baby throughout his life Practically: We needed to drive and stay over for two nights at the treating hospital x4 times and so this was uncomfortable for him for the last trip because he had increasing secretions and he needed to be in a lie flat cot on his side in the car.

2. Impact on you / your family

Type 1, treatment started <7 months, 5-7 injections

Seeing his development is an amazing emotional boost through such a hard time. Without it I think he would have died from respiratory failure.

Type 1, treatment started age 8 - 12 months, 5-7 injections

I think the treatment has had a huge emotional impact for us as although it's not a cure it has offered us hope, hope for a better future for our family. Her gross motor skills have improved slightly and this makes it easier to play. Also as the disease has not progressed we are not dependent on a ventilator so we are still able to go out as a family. It is stressful attending the treatment because as a parent you do not want to put your child through a painful procedure but I feel the benefits far outweigh this.

Type 1, treatment started age 8 - 12 months, 5-7 injections

...Without this we were given no hope I wake up with more and more fight and goals everyday this really is an amazing drug/treatment. Physio and games are so much more fun now she wants to join in she's the happiest strongest little girl since receiving this. Myself, the rest of our family my physio my occupational therapist and language and speech therapists are all in their element they are now giving her tasks and equipment we'd never thought we would have the option to give her...

Type 1, treatment started age 8 - 12 months, 5-7 injections

Is giving me hope. It makes me strong, because I know I do have more time with my child.

Type 1, treatment started age 13-24 months, 5-7 injections

We live in hope now, no longer fear, for what her future holds.

Type 1, treatment started age 13-24 months, 5-7 injections

This has completely turned our lives around...We were told to enjoy our time left with our child at point of diagnosis and before treatment had become available which was simply heart-breaking. Life as we knew it stopped. Numb with pain and filled with fear we were unable to work/sleep/deal with normal day to day life. However now I'm witnessing first-hand the benefits of nusinersen I'm simply filled with hope for my child's future. This has had such a positive turnaround for our family, myself, my husband, siblings, grandparents. I feel like I'm no longer waiting on a ticking time bomb, but now look forward to my child's future.

Type 1, treatment started <7months, 11+ injections

When your child is unstable and having frequent hospital / ambulance admissions this is very draining both physically and emotionally on the whole family. We are more relaxed and able to enjoy day to day life and activities so much more now. SMA is very tough on you as a carer / sibling, but with his stability and health being so much better we feel a lot more happier as a family.

Type 1 / 2 treatment started 13-24 months, 0-4 injections

It gives us hope. It has made looking after our child easier as she does not need feeding or respiratory support

Type 1 / 2 treatment started 37 months +, 5-7 injections

She has had less chest infections and her bipap pressures have decreased, so less intensive physio
The leaning is kind of a pain she has had more accidents in her chair due to leaning

Type 1 /2, treatment started 37 months+, 5-7 injections

Everyone is more positive, and can see that it is possible to move forward, not just slide backwards as previously.

Type 2, treatment started, 0 - 4 injections

Get inspired by gradual improvement, takes pressure off emotionally by having to do less in terms of care and knowing that the degeneration could be halted. Reaching stability is essential for future further improvements by new drugs. It is no longer about what my daughter could do and no longer can. It is about what she could not do before, but can do now. This is very exciting.

Type 3, treatment started age 12 years, 5-7 injections

Spinraza is having an enormous positive-effect on our family. Physically: Before treatment he began to fatigue extremely easily. He was too heavy for me, but my husband would carry him upstairs if he had become too tired to crawl up/down. With Spinraza he can manage walking up/down stairs. When he fell/collapsed before treatment, he required assistance to get up either by myself/husband/chair etc. With Spinraza, he can now get up independently from the floor & isn't falling. Before Spinraza, he needed help dressing/putting socks/shoes/splints on. Since Spinraza, he can do this. With Spinraza treatment, he is stronger/with more stamina which has led to reduced physical demands for carers. We are less anxious about his condition as he isn't falling and is doing so much better..... My husband just started a new job in January 2018-he felt he could take on the new challenge. I too, in January 2018, had a promotion. We wouldn't have taken on these new roles before... Before treatment, we knew that at least one of us would have to quit our job to care for him as he deteriorated. We can continue our careers & progress within our roles....Reduced caring responsibilities mean that family life has become easier and happier for all of us & of course most importantly for him....we're amazed at our son's response to treatment changing our lives and futures for the better.

Bereaved: Type 1, treatment started < 7 month, 0 - 4 injections

Physically: Nil. Emotionally: There was some delay in a decision being taken as to whether we could access the treatment and we initially received a letter stating we were not allowed. This was stressful because in his deteriorating condition, time was critical. We prompted and chased for our baby's paperwork to be transferred to apply for this treatment and we wrote to the CEO of our area hospital. We visited the xx Children's Hospital and hand delivered a letter to the Acting CEO and that day the decision was changed. This challenge was stressful, upsetting and frustrating. We experienced great

distress as a result of conflicting expectations of the likely impact of the drug set by teams in two hospitals. One told us that the drug would slow or even halt the decline. Whereas the other told us that it could reverse the process which would allow him to reach milestones and that he would sit up and possibly even walk. This gave us hope, joy and relief, but later grief when these milestones failed to materialise. Practically: We both had long drives, overnight stays and time away from home and more time off work causing even more disruption. We needed to have relatives travel to our house and stay over to look after pets which added to disruption and made living with SMA slightly harder. We considered the practicalities of moving (e.g. USA or treating hospital area) to allow us access to Nusinersen when we could not access it due to living where we do.

3. Expectations for the future

Type 1, <7 months, 5 - 7 injections

I expect the treatment to continue for his life - he continues to improve month by month and we expect him to walk by himself one day.

Type 1, treatment started age 8 - 12 months, 5 - 7 injections

I hope that the treatment will allow our daughter to gain more strength, I hope that she may be able to achieve supported sitting to use a wheelchair and offers us more time together as a family.

Type 1, treatment started age 8 - 12 months, 5-7 injections

I am hoping that she will become stronger and stronger with every dose of nusinersen.

Type 1, treatment started 8 - 12 months, 5 - 7 injections

I'm hoping we can improve respiratory more but from what I can see this drug is helping more upper body I'm hoping to see gravitational movements and head control she now has the fight to try. I'm excited to see what future treatments will bring it really is the wonder drug. I'm hoping this will carry her through until they find a cure....

Type 1, treatment started age 13 - 24 months, 5 - 7 injections

We would be thrilled with just stability. We never expected to see the gains we've seen at the outset of treatment

Type 1, treatment started age 13 - 24 months, 5-7 injections

Each treatment only makes my child stronger fact. Albeit small improvements at times, any improvement with this condition is a miracle. I expect future treatments to create slow and steady gains. I strongly believe physio also plays a huge part too so regular exercise will be required. I'm unsure to the final extent of my child's progress but I strongly believe she will at some point be able to hold her own head and as she's gained some strength in her arms I believe this will make a huge impact to the quality of her life. I'm quite realistic and I don't expect my child to perhaps walk but

sometimes I wonder will her type 1 condition now become more like a type 2 - and for this I would be very grateful.

Type 1, treatment started < 7 months, 11+ injections

Stability is the key. No deterioration. We as a family work hard with him to increase his physical strength and our aim for this year is to get him moving confidently in his power chair and to hold his head.

Type1 /2, treatment started age 37 months +, 5-7 injections

We would really like to improve her swallow and reflux

Type 1 / 2 treatment started age 13 - 24 months, 5 - 7 injections

For gradual improvements to continue

Type 1 / 2 treatment started age 37 months + 5-7 injections

Type 1 and 1/2 children are described as having "life limiting condition" due to respiratory complications. With Spinraza helping with respiratory functions, better life expectancy is the main advantage. Also with fine motor skills increasing, we hope for more independence for our daughter, like ability to feed herself and more confident use of cutlery, being able to write better, reach to the top of the keyboard, turn the page in the book etc.

Type 2, treatment started age, 0 - 4 injections

Improvement by prolonged exposure

Type 3, treatment started age 12 years, 5-7 injections

....This treatment is lifechanging for him with significant ongoing physical improvements. This has positively correlated with his emotional wellbeing where he now looks forward to the future where he can fulfil his goals & isn't fearful of being trapped in a disabled body... With Spinraza treatment at home in England he will need to access other health services less over time as his SMA disease progression has halted & his abilities are improving. Yet without treatment, over time he would require increasing Health & Social Care services. His health needs would become increasingly complex and costly to health & social care services as his abilities degenerate & associated sequelae onset with increasing suffering for him.

Scottish Survey December 2017 – January 2018

Treated with nusinersen

Three replies were from parents living in from Scotland; four were from parents living in England

- **Parent of child with SMA Type 1 / 2, now age 2 years 2 months. Treatment started August 2017 at age 22 months. By 31/12/17 had had 4 doses**

Was only declining but we have now seen improvements since having the drug. More strength in arms, legs and better head control

We hope to see gradual improvements which are ongoing. We hope one day our child can sit independently and eat lumpier foods

For a condition which only worsens this drug is a real hope. To someone who cannot move some improvement is better than nothing. Only families/people in that position can understand what that means. It could mean more years of life

- **Parent of child with SMA Type 1 / 2, now age 10 years. Treatment started April 2017 at age 9 years. By 31/12/17 had had 5 doses**

Improved fine motor skills, improved overall movements, improved respiratory functions, improved cough, less chest infections.

Reassurance that something can be done to stop deterioration. Improved respiratory function and cough, improve motor skills and overall movement. Less chest infections.

Nusinersen gave us hope. With less chest infection (which can be life threatening), life is less stressful. It gave us motivation to continue therapies and improve our child's chances to get better. It's wonderful to see improvements for the first time, after so many years of decline in our child's health.

- **Parent of child with SMA Type 1, now age 14 months. Treatment started June 2017 at age 7.5 months. By 31/12/17 had had 5 doses**

My child is now 14 months and can sit unaided for around 30 minutes, can roll from back to side, eats orally, breathes on own with no intervention at all, now uses a stander for 2 hours per day

We as a family are so grateful for nusinersen, we had prepared ourselves for how sma type 1 robs kids of life etc our child has took a whole different path and amazes everyone who sees how well they are doing

My expectations are that my child has the potential to walk, live a relatively normal life

- **Parent of child with SMA Type 1 /2, now age 3 years 8 months. Treatment started February 2017 at age 2 years 10 months. By 31/12/17 had had 6 doses**

It's completely changed our lives. Before nusinersen my child had started the cycle of going in and out of PICU. Every illness resulted in a left lung collapse. We'd always go into hospital with a pretty hefty PICU stay, never the ward. Since starting we've had a couple of very relaxed ward stays as our child needed a small amount of oxygen through their bipap at night. It's been 6 months since our last chest admission and we've handled a few horrific coughs at home, these coughs were far worse than the ones that had our child intubated. They now have a proper cough. If you'd have asked us this time last year how we thought we'd be doing now we probably would have thought we'd lose our baby this year. Now we have a strong, confident amazing child, who is whizzing about in their power chair with far more strength to play. They have head control, regained most of their swallow, can lift some toys, almost sitting, can move legs, can cough, shout, cuddle all the things they couldn't before.

Obviously, a level of worry will always be there, as it's not a cure but we make sure we follow all protocol; we are very strict with bipap, chest physio and general physio which is incredibly important. Nusinersen works alongside protocol it's not instead of. Since starting the treatment and seeing our child's progress, for the first time we've allowed ourselves to look to the future, to think what we'll do in the future, to think about them growing up. Before Nusinersen everything we did was touched with sadness, in the back of our minds we knew that at some point we'd lose our child. Every snuffle or cough was terrifying. It's no life constantly living in fear, wondering if this is the last birthday or Christmas.

There needs to be protocol to go along with Nusinersen, bipap physio etc this will give patients the best chance... it needs to be led by hospitals with amazing respiratory departments.

- **Parent of a child with SMA Type 1 now aged 14 months. Treatment started January 2017 at age 13 weeks 5 days. By 31/12/17 had had 6 doses**

They now have full head control, can sit unassisted for up to 15 minutes with trunk being supported by a lycra suit to help prevent curvature of the spine. Can now move head freely from left to right, and hold in a neutral position. Can easily track people round the room. Can lift arms fully above head when in a slightly reclined seating position and can lift them to shoulder height when in an upright seated position. Can now lift and hold much heavier objects than could ever do in the past. Has increasing hip and leg movement and when lying on back, can lift leg off of the ground and is starting to initiate a roll to the side. With encouragement, can roll right on to side (both sides) however is still reluctant to carry this on further so that is lying on tummy.

Emotionally - We feel that our child is achieving all emotional milestones that would be expected of a typical 14 month old. Practically - can now sit in a supportive chair and play with age appropriate toys, can interact with sibling and peers, sit independently for a short period of time on own and is aware of surroundings and family. Reacts to familiar faces, points to things seen and wanted and is very vocal in babbling.

Physically - At this moment in time we still have to assist with lifting, carrying and transferring a 10kg baby. This is physically demanding and tiring and has resulted in constant back ache and shoulder ache and RSI.

Emotionally - Seeing the amazing progress that our child is making means that we have so much more hope for his future.

With continued nusinersen treatment it is our hope that our child's respiratory system and swallow may strengthen to the point that they have a safe enough swallow for some solid foods to be introduced.

Without the drug nusinersen, the historical nature of this disease means that most babies born with SMA type 1 will most probably die before they reach their second birthday, due to the degeneration of muscles throughout the body but crucially, those around the respiratory and feeding system. Therefore, without the interventional use of this drug, these children will ultimately be condemned to certain premature death.

For those children with SMA Type 2 and 3, I believe the drug nusinersen would absolutely enable them to regain some independence and allow them to regain lost skills or learn to do things that they never could before e.g. a twelve-year-old child who sits daily in a motorised wheelchair, unable to lift their hands beyond their own chest height, could, with nusinersen, be able to lift their hands so that they can then scratch or blow their nose, comb their hair, itch their eye, reach to switch on/off a light switch etc.

The last thing to be considered by the NHS in Scotland is the continued and increasing cost of the palliative care that is required and given to patients all over the country with SMA, versus the cost of a drug that would alleviate some of that burden to the NHS as patients become more mobile and require less physical support. Finally, with other pharmaceutical companies such as Avexis, researching and producing gene therapy drugs which could provide a cure for SMA, I feel that the nusinersen drug will be replaced in time with a complete cure.

- **Parent of child with SMA Type 1, now age 16 months. Treatment started October 2017 at age 13 months. By 31/12/17, had had 4 doses**

Increased movement in legs and arms. Is finding it easier to play and interact with others. Think breathing may have improved also. Still lack of head control. May have caused some vaginal bleeding. Happy as evidence of increased movement. Only ever needed an overnight stay after treatment for monitoring. We have been accepted to have the first of the maintenance doses and we'll be reviewed after this 5th dose.

- **Parent of child with SMA Type 1, now age 2 years 6 months. Treatment started June 2017 at age 2 years. By 31/12/17, had had 5 doses**

Emotionally - has had an impact where our child feels sense of achievement at the improvements they are making, feels more independent & has given a sense of hope that they can do more & more. They feel more included as is able to join in with more toys & games & just in general has more energy so is upbeat & happier.

Practical - able to do simple tasks like feeding self more easily, has more options for play, can now help to wash self & can help with tasks such as baking & helping with practical tasks in the house ... feel more included & part of the family so not missing out on anything.

We hope to see continued improvement & halting of the muscle wasting for our son.

We feel this treatment would be a life saver for people with sma & with the improvements we have seen in our child we feel this treatment should be considered for all types. To see milestones being reached that were never possible before is an incredible achievement for our children. Even to have the ability to sit up, feed yourself, lift & play with toys gives our children so much more opportunity to enjoy life than they would have without this treatment.