

**Appendix 2.
Survey Jan – Feb 2018
Impact of SMA Types 1, 1 / 2, 2
ages 0 – 17 years
Biggest Challenges**

Type 2 age 0-2 years mother

As a parent of a nearly 2-year-old with type 2, Emotionally: Massive impact knowing that your son has a degenerative condition and potentially a reduced life span. The Spinraza success stories that are so easily accessible on social media also have a significant impact, on the one hand there is genuine hope on the other a desperate feeling of helplessness and injustice that this isn't open to all in the UK. Physically: This will only become more of an impact as he and us as parents grow older. Practically: Very concerned about how we will manage the physical load as he grows and have the appropriate housing/equipment to give him the best quality of life

Type 2 age 0 – 2 years father

Spending enough time with my child as a working person and keep myself emotionally stable is the biggest challenge.

Type 2 age 0 – 2 years aunt/uncle

The biggest challenges relate to deterioration of muscle strength and the fact that things often go backwards for a child with SMA. Emotionally, this has a massive impact on a child, who is at least as, if not more intelligent than other non-affected children, as they are already aware of their limitations and struggle to understand why they can no longer do the few things which they were able to do a few months before. Practically for parents supporting such children, the constant need for adapting care to take changing needs into account can be physically, emotionally and financially draining.

Type 2 age 0-2 years grandparent

The need to adapt the house to cope with wheelchair, wizzybug and standing frame. Access to the upper floor and to the garden and its subsequent use. Paths and patios, for example, need to be adapted. Sleep difficulties can make for reduced energy during the day. As he grows older there will be problems with access to bathroom and bedroom.

Type 2 age 0 – 2 years parent

The major impact for our son is in his physical ability to move. He cannot crawl, stand or walk, and has very restricted movement and strength in his body. Additionally, his breathing is affected as he has little strength in his torso, which directly affects his lung capacity. His breathing can be shallow and quick.

Type 2 age 0 – 2 years grandparent

*Increasing muscle weakness is heart-breaking to experience for both child and family. It is becoming harder for my grandson to press buttons on his electronic toys and lift things. *My grandson's bones are becoming brittle and he has experienced 3 fractures in 3 months. It set him back because he could not continue with his OT exercise or attend hydrotherapy

sessions. In a normal healthy child these would not have occurred. *My son and daughter-in-law get virtually no sleep. *We dread him getting a chest infection. *It is frustrating for him and heart-breaking for us to see him grabbing his trousers to physically lift and move his legs to try and reach a toy. *It is difficult for my son and daughter-in-law to work and find carers now that his needs are growing. Transferring him from chair to changing mat to pushchair etc is not easy now he is heavier and bigger. As ageing grandparents and part time carers this is hard work and takes its toll on our health. *Both parents are now experiencing back pain. *Changing mats in shop toilets are no longer appropriate which makes shopping and social meetings with friends more difficult. *Travel on public transport can often be an issue with a child under the age of 3 with SMA because they look normal and are not in a wheelchair. It is assumed that he is able bodied and expected to fold the pushchair to make way for wheelchair users. I have been verbally abused for not doing this! *Although the council have been generous in their disability grant it is nowhere near enough to pay for the adaptations needed to the house to accommodate his growing needs.

Type 2 age 0 – 2 years parent

Uncomfortable at night, sleep study suggests it's not down to low oxygen, high carbon monoxide, more likely to be lack of strength to reposition himself.

Type 2 age 0-2 years father

The hardest part of SMA for me is the regression. We all look forward to our children hitting their milestones and we celebrate these occasions and then when they get to a milestone and don't progress past it, you start to ask questions and speak to doctors. Then comes the diagnoses, which is hard enough, but to then start having to watch your child lose his greatly achieved milestone it's heart-breaking, you can't explain to him why he can't do that thing he was doing two months ago. Our children, some are only young like our son, however they look at the other kids that come to play and you can see them wondering why they can't stand, walk, crawl or jump. You can see in their eyes they want to be doing it, they try to do it. How do you explain this to a two-year-old? As parents there isn't anything you wouldn't do for your children, if I was given the option to give up my ability to do these things just so he could do these things I would do it in a heartbeat. But to be in position where there isn't anything you wouldn't do but there is nothing you can...it's the worst. You feel like you've failed.... you've failed your baby and he's only 2.

Type 2 age 0-2 years father

Living with SMA is incredibly challenging. SMA has taken my son's ability to move independently. He will never be able to crawl, stand or walk without a successful drug intervention. Long term his lack of strength will affect his ability to live an independent and free life. It might affect his ability to make friendships and he may become isolated at school. He is extremely vulnerable to (relatively) minor chest infections, colds and viruses which, due to his poor breathing ability, can become extremely serious very quickly. Last year for example he was admitted to A and E twice and both times spent at least a week on the High Dependency Unit ward. Emotionally it's very hard on my wife and I, and on our wider family. We worry about the smallest cough or snuffle, and are constantly watching him for signs of a deterioration in his condition. This is exhausting, and although we are determined to live a full and happy life, the worry is always there. Practically our house is full of medical devices and equipment. If we want to go on a trip overnight there is an assisted cough machine and a nebuliser to take, as well as a sleep aid and maybe a specialised chair. Our 'normal' is very different from most

peoples', yet we are a strong unit and feel blessed to have such a beautiful little boy in our lives. We wouldn't change him for the world.

Type 2 age 3-4 mother

My son has been admitted into Intensive Care with lung collapses and consequently been intubated 7 times and he is only 4 years old. These awfully traumatic episodes had to be managed whilst packing up our house for a 5-month renovation to make it accessible, managing the build, making sure our teenage son was being cared for and juggling hospital around a full-time job. When he is not in hospital we are petrified of him becoming ill again and try to live life to its fullest whilst protecting him from numerous bugs. This unfortunately means long term absence from school when the children in his class have coughs/colds/gastric illnesses/viruses. He misses out intellectually and socially. The care we provide for him is intense yet does not mean that he will not become ill again....we live in fear. We do not function as a 'normal' family would. My husband and I do not go out together as one of us has to stay with him and we do not go on family holidays as 1) he has a lot of equipment and 2) we are frightened to leave the area in case he becomes unwell and may need urgent intensive care treatment. SMA has affected us all and our lives will never be the same again.

Type 2 age 3-4 years parent completing

PHYSICALLY - I am restricted a lot in what I wish to do and heavily rely on my carers such as my mother and father. Simple things such as toileting, cleaning myself dressing myself I am unable to do so without support. I feel extremely sad that as I get older I have to rely on someone else support to dress, shower and use the toilet. I wish I had the opportunity to do things by myself and not rely on my parents. I am 3 and want to explore and have fun, play however due to my physical restrictions I am unable to enjoy and play with my friends how I like and feel like a burden sometimes on my social group as they have to help and assist me in my play. It makes me sad and sometimes cautious in how I play. **EMOTIONALLY** - I feel very reliant on my family and my peers to help me get around and that can make me feel inadequate and burden to my loved ones. I am extremely anxious because of my limitations however, deep down I am adventurous and love to new experiences that my family facilitate for me. I have to ask permission to do things I should be able to do such as help in going to bed, undress my clothes, clean my clothes etc and makes me upset that I am not able to do these simple tasks by myself. it's very frustrating and sometimes makes me angry with the situation. **PRACTICALLY** - the basic of human right to be able to wipe my bum I am unable to do due to the lack of muscle strength I have and that brings me down. Drawing and colouring is something I truly enjoy but due to the lack of hand strength I have it is hard and tiresome when done for a short period of time.

Type 2 age 3-4 years mother

The biggest challenges I face as a carer with a child with SMA are many. Physically, I am restricted in where I take my daughter and how I deal with her needs. I have to make sure that the place I am going to is suitable and I am able to hold her for a little while to get her to where we need to go. I get extremely tired in trying to help my daughter have a childhood and do things with her that a normal child her age is able to do. I feel pressured in many ways to deal with her needs without any support from outside help. My daughter is now nearly three and a half and is extremely demanding with her needs which is tiresome and incredibly overwhelming. I also feel a sense of extreme guilt when I am

unable to take her to places where I am not able to carry her around for long periods of time and also going to friends and families houses where there may not be accessible and all on one level. I sometimes feel very tired at night because of the day to day roles I have to do with her including toileting cleaning feeding playing etc. Also, I feel responsible for her physical therapy needs as the help and advice I receive is limited and solely depends on me fulfilling them and I feel guilt when I lack in doing it daily or when recommended. I sometimes feel part of my life as a person has been taken away from me because of the responsibility I have to take care of my daughter and make sure all her needs are met before mine. The lack of independence makes her completely reliant on me and upsets me to see her struggle in the most basic activities such as colouring playing etc

Type 2 age 3-4 years grandparent

My grandson is unable to walk or stand and can sit only in with support. He is susceptible to serious respiratory problems initiated by the common cold/hayfever etc. This leads to frequent emergency admissions to PHDU and PICU for up to 5 weeks at a time - the stress placed both on the child and, probably more so on the parents in these dangerous situations is immeasurable. His swallow is exceedingly poor necessitating feeding direct into the stomach, nil by mouth. It is obvious that these problems necessitate 24-hour care and my daughter is unable to work. It is proving difficult to obtain official help for my daughter - instead she relies on her partner during his non-working hours and some unskilled help from myself.

Type 2 age 3 – 4 years parent

My daughter is 3 and a half and struggles a lot with drawing and colouring using normal pencils. She gets extremely tired when using her hands for drawing, writing, playing with play dough etc. She gets quite upset that she cannot do it for a long time and frustrated. Help with getting toys and to play with them Support in positioning, if she's laying or falls over she needs help in sitting back up help with turning over and lifting her legs

Type 2 age 3-4 years mother

As the main carer I continue to work as the cost of SMA is huge and therefore I do not feel in a position to give up. However, when our daughter is unwell, particularly from September-March, work can become very difficult as she is unable to attend school/nursery due to the increased risk to her health (mainly surrounding respiratory problems) and there are very few others who are equipped to deal with her medical needs. For our daughter herself, her lack of independence through needing support for absolutely everything can be very challenging. She cannot be left unsupervised as she is unable to access her surroundings on her own. This makes her being able to socialise or integrate with her peers more difficult (she lacks arm strength and dexterity in her fingers even when she is able to follow them in her powered chair) She becomes very frustrated at not being able to complete the simplest of tasks (do zips, use cutlery properly, brush her own teeth, not being able to wipe herself after toileting) and she will often get angry or cry at these barriers.

Type 2 age 3-4 years mother

For me (the unpaid carer), the biggest challenges are: - Lack of sleep (I wake up 8-10 times a night, every night, to turn my son) - Emotional distress at seeing my son's strength deteriorate in front of my eyes, despite everything we do to keep him as strong and as well as possible. He is now almost 4 years old. In 3 years, he has lost the ability to raise his arms, crawl and roll. He can no longer even sit

for longer than 5 minutes without the support of a back brace] - Emotional distress at knowing that there is now, finally, a treatment available that can help him and change the trajectory of his life, but we cannot access it - Physical pain in my back from lifting and transferring my son, and constant fear about what will happen to him/ who will take care of him if anything happens to me For my 3 year old son (the person with SMA), the biggest challenges are: - Fatigue - Frustration and sadness about not being able to do all the things he sees his friends doing

Type 2 age 5-12 years father

Its 24hr care for the person with SMA. Your whole world, time and place revolves around them. It can be physically draining, emotionally upsetting. Fulfilling their need for personal care, clothing, toileting, cleaning the wheelchair mess and track during the rainy days, their educational needs and support, having them entertained in terms of their normal childhood activities, getting them new friends are all very difficult. Toileting outside her home or school is very difficult to get access unless you are in airport etc where you get some decent changing or caring facilities. Finding the time to spend as a child, going to park, going to cinema, or shopping, travelling it is all a biggest challenge. Can't attend everyone's birthday party or sleep over at their friends' place, as no accessibility or toileting facilities for the child to go to their friends' place. These are basic childhood activities and part of growing experiences which child misses due to her limitation in her mobility...

Type 2 age 5-12 years mother

The emotional impact on the other siblings. Financial and emotional impact on the family. Social impact on child with SMA. Issues with accessibility for the child with SMA. Unable to access parts of their education. Financial demands on buying WAVs and equipment. Difficulties accessing treatment. Difficulties getting correct equipment for child.

Type 2 age 5-12 years mother

Keeping our son well throughout the winter proves challenging. The slightest cough, sniffle has me on pins for fear it leads to pneumonia or a collapsed lung. The emotional battle is pure torture and my anxiety is ridiculous. I live in constant fear that his body may one day be too weak to recover and that thought tears through my heart. My physical health is poor due to lifting, not sleeping or eating enough.

Type 2 age 5-12 years mother

I am a qualified professional (accountant) and would love to return to work full time. I worked until just 2 weeks before my daughter was born, but have been unable to work since (so for the past 12 years). I am unable to sleep at night as I have to roll my daughter frequently (every 45-60mins or so) as she has a lot of pain in her hip joints. That means I have to try and sleep during the day, though most days it doesn't happen. Also, due to suppressed immunity my daughter gets ill quite often, which means she would have to stay at home so as not to add any more complications (unlike an able-bodied child who can return to school quite soon). All the hospital appointments, treatments, surgeries, etc take up a lot of our time. All of the above impacts on my day and the ability to work or even function properly sometimes. When she is home from school in the evenings, during school holidays and weekends, I have to do a lot of physiotherapy with her (conditioning, strengthening, stretching, chest physio, secretions, etc), that takes up a lot of the time when parents of able-bodied kids would normally get on with household chores, extra work, cooking or spending time together as

a family. That means I have to do all of the household chores, shopping, cooking, etc while my kids are at school, because as soon as my disabled daughter is home she needs my help with everything (bathing, toileting, physio, getting dressed, doing homework, etc). My able-bodied daughter often feels neglected (and currently received counselling) and I am constantly torn and feel guilty. My marriage to the girls' dad ended a few years after the diagnosis and I think it was in main part due to all the extra difficulties we faced. Basically, SMA has had a huge negative impact on the whole family in every area of our lives - financial, emotional, marital, personal, self-fulfilment and physical health.

Type 2 age 5-12 years grandparent

The lack of joined-up thinking on the part of the medical, allied and support professions. All seem keen to promote their speciality - surgery, orthotics, respiratory care etc - and not see the overall picture / implications for the patient and the effect this has on the carer. Advice can conflict. The daily battle to provide a 'normal' life for the family. Time is taken up with what should be unnecessary issues - e.g. provision of adequate one-to-one care at school, where again, there is no oversight, planning or even co-operation about, for example, school trips or absence of staff trained in the care of the patient. This can be emotionally draining for the family.

Type 2 age 5-12 years aunt/uncle

It is very difficult to watch a child's condition deteriorating. It is a huge challenge as it is to have no use of your legs. As he gets older and bigger the strain of moving and carrying him means more adaptations are needed in the home and less places are accessible. Joining in at school is becoming more difficult. Not being able to go to friends and family's homes. Needing to be turned in the night. Struggling with weight gain. Watching him become less balanced, not being able to sit unaided. Everything getting weaker.

Type 2 age 5 – 12 years grandparent

My daughter is the mother of a SMA type 2 daughter who is in constant pain with permanent dislocated hip joints, this causes my daughter to have to tend to her child many times through the night to turn or "Click" her hips. She is double incontinent which in itself is a problem when going out anywhere as there is never a convenient changing facility other than baby changing facility. My daughter has a back problem which is now being aggravated by constant lifting of a 10-year-old child who has been in a wheelchair from the age of 2.1/2.

Type 2 age 5-12 years mother

The progressive lack of being able to do things for himself. Now reaching teenage years & having to become more dependent on me. Not being able to accomplish the most trivial of tasks like sitting up by himself. The sheer lack of help from local healthcare i.e. physiotherapist leaves us completely on our own & making up things as we go along.

Type 2 age 5 – 12 years aunt/uncle

Exercise is a big challenge for my Nephew as he is in decline and unable to sit up or turn himself over in bed at night. He lacks strength in his upper body and has previously has some strength there. He used to be able to go horse riding which was great for his core muscles but sadly is not able to do this anymore. His Mum struggles to be able to take him out on her own as she isn't strong enough to carry him anymore as he is growing and so they find that they are mostly homebound.

Type 2 age 5-12 years (parent completing)

My biggest challenges are trying to build my Lego but my fingers are not strong enough. My mum helps me with everything I want to do but I know she struggles lifting me as she has a really bad back. It's a struggle to bath me, and get me dressed as I cannot lift up my arms to help. It sometimes takes a long time to get my shoes on ready for school because my feet are very delicate. I sometimes feel sad when I don't get to see my friends if they have a cold because it means I could end up in Hospital really poorly. My Mummy doesn't really get to see her friends much or go out as she worries about me too much.

Type 2 age 5 – 12 years mother

The biggest challenges for our daughter is not being able to get around the house without help from us. Due to the muscle weakness she also has challenges completing day to day tasks such as cutting up meat and opening drink bottles.

Type 2 age 5-12 years mother

1) The availability of Changing Places Toilets. 2) Daily Physiotherapy 3) School Holiday cover 4) Hospital appointments / Missing School 5) Integrating with School Friends 6) Coughs and colds 7) Complications from Spinal Surgery

Type 2 age 5-12 years mother

The emotional impact on the other siblings. Financial and emotional impact on the family. Social impact on child with SMA. Issues with accessibility for the child with SMA. Unable to access parts of their education. Financial demands on buying WAVs and equipment. Difficulties accessing treatment. Difficulties getting correct equipment for child.

Type 2 age 5-12 years young person

Not being able to do the things other people take for granted - picking up a drink to sip it, rolling over in bed, drawing with a pencil, getting in and out of bed, privacy in the toilet and with personal care, to name but a few.

Type 2 age 5-12 years young person

I can't walk, I can't reach out things I wanted, I can't use steps, I can't go and visit my friends home as steps in their homes. I won't get enough things to play with my wheel chair if I go to park

Type 2 age 5-12 years father

EVERY daily task is a challenge for both the affected person and the carer. it takes considerable amount of planning and assistance from carer, before a simple task can be carried out i.e. helping with homework, picking a dropped item from floor, cleanliness and hygiene, going out to a park, dropping/collecting from school, toileting, feeding etc. it is physically exhausting routine where carers are 24/7 on their feet with little time for their selves. It is extremely hard for families who don't qualify for all NHS related services/benefits and little assistance with mobility equipment.

Type 2 Age 5 – 12 years parent

Needs help with adjusting clothes, reaching out things in and around the house. Needs to keep house warm, heating is on most of the time from Autumn to Spring time.

Type 2 Age 5-12 years parent

My son has very recently had surgery for hamstring lengthening. This is due to the excessive tightness which have occurred in his knees meaning he now hasn't been upright in a year. Regardless of the physical implications of this emotionally it has been extremely hard on him. He is only 6 and now has to spend the next 6 weeks using a manual wheelchair in which his legs can be out straight in gaiters for 24 hours a day. This is causing intense pain and upset. He is unable to return to school due to lack of funding to ensure that he has the right equipment to use the toilet. It takes 2 people to lift him after this surgery we understand there is no funding for any outside agency to come and help with the lifting meaning it is down solely to me as his mother to try and meet his care needs.

Type 2 age 5 – 12 years grandparent

The main problem is a failure to integrate care both within the NHS and between NHS and social care. This shows itself in everything from getting a supply of continence products to a year-long delay in replacing a broken and outgrown wheelchair. Individual elements of care within the NHS are usually good, apart from the wheelchair, but the complete failure to integrate the care and make it patient not institution focused. Each hospital visit involves my daughter in 165 miles of driving and visits occur about 6 times per month, sometimes three days in one week. Despite complaints the hospital makes no attempt to coordinate visits. Quite apart from the stress on my daughter the loss of education time is considerable.

Type 2 age 5-12 years mother

The biggest difficulty is the lack of physical independence for my son. He is very independently minded and wants to be as autonomous as possible and is frustrated when he can't be. He doesn't want a carer with him at all times but he needs to have one as he is unable to do most things himself.

Type 2 age 13-17 years young person:

It's so hard to always have to rely on others for almost everything. Both my parents are always tired because of the sleepless nights. They are emotionally drained - it must be so hard to see your child struggle and I try to ask the minimum.

Type 2 Age 13 -17 years young person/parent jointly

Going to school in a taxi with an assistant; requires full assistance to call lifts, open doors, get in a bus/taxi. Full support to access resources at school and home. Full time support to assist with moving arms to scratch an itch, turn on wheelchair, pick up things. Completely dependent on others at all times to perform any basic task requiring movement.

Type 2 age 13-17 years young person

I cannot do the simplest things on my own: lift my hand to my face, pick up a cup with water, keep my head upright. Depending on someone (an adult) all the time, makes it difficult to be independent as every boy my age wants to be. I cannot go to meet my friends on my own, I cannot go to their houses (not accessible), I cannot hang out with them without having everything pre-arranged so a carer is present. These are very few of the things that are practically difficult for me to do and lead to me being sad and emotional quite often. Similarly, my parents have to look after me 24/7 during holidays (no support then) while taking care of my younger brother. There is also no support during

the weekend and therefore if my parents have any energy left to do some activities with me and my brother, they always have to be suitable for both of us - the common ground is very limited (and boring!) Quite often, one of my parents stays with me, while the other is with my younger brother; we would much prefer to do things together as a family, but this is usually not practical. During the week both my parents work full time and as soon as they get back home they take over my carer in looking after me. This is both physically and emotionally demanding for everyone involved. I really want to be more independent, I do not want an adult with me at all times (even in the toilet), but it is impossible for me to practically function otherwise. I'm smart, top of my year in my (mainstream) school and worried about what the future will bring, as I want to go to university and get a job.

Type 2 age 13-17 years young person

My unpaid carer must do all living needs for me, e.g. assisting with meal times, assisting with turns during night time and any time of the day when I am in bed. Personal grooming, like bathing and toileting. I do need re positioning when I am in the wheelchair. I want to be more independent and do some stuff for myself as I see how hard it is for my mother.

Type 2 age 13-17 years young person

Every new day is a new challenge for me and my family. Every breath is a challenge, every dress up is a challenge. We are trying to keep the motivation, but it is very difficult when we see how it is getting worse my condition. Do you know how it is when you cannot scratch your head when it is itchy? Do you know how it is when you are hungry or thirsty and you cannot even hold a mug in your hand?

Type 2 age 13-17 years mother

Lack of independence, being reliant on other people to help them all the time e.g. going to the toilet, getting up and going to bed, getting dressed. Constant pain of back and dislocated hip. Lack of sleep because of turning required.

Type 2 age 13-17 years mother

My son doesn't think he has a future like his brothers, doesn't believe anyone will employ him or that he will live independently. Both we have always told him he can do but his rapid loss of ability has taken away his confidence and self-belief. He has become more isolated, doesn't want his friends to see that he can't hold his head up if it falls forward so avoids putting himself in a position where he might need to ask for help and has slowly been pulling away from going out. Stopped enjoying going out for family meals as doesn't want us feeding him in public, he does now have a peg fitted but can eat foods he really likes in small amounts - will only eat at home now. As a family we are further restricted in what we can do and enjoy together, as a couple we rarely do anything together and don't see that changing