

Appendix 3.

Survey January – February 2018

Impact of SMA Type 1, 1 / 2, 2 - ages 18+ years

Biggest Challenges

Type 2 age 18-25 years young adult

My lack of strength. I wish to be able to do many things for myself, but I struggle due to lack of strength. Simple things like lifting my arms up to put my headphones in, or even put my glasses on. I have to use two hands to lift drinks up and I take about 25-30mins to finish my meals as I struggle to use knives and forks, and swallow - and by this time my food has gone cold. Emotionally this plays a part on me too - I often feel embarrassed that I can't do the simplest of things.

Type 2 age 18-25 years young adult

My biggest challenge is balancing my social life and hobbies with the limits of my disability. I quite often push myself too far and suffer exhaustion and fatigue.

Type 2 age 18-25 years mother

Everything you do or want to do takes maximum effort. SMA sufferers are very intelligent and require stimulation and variety in their daily lives, this is achieved with a great deal of effort and support. Parents have to be nurses, physio, OT's, carers, therapists and parents all on a lack of sleep and reduced earnings. You live in constant fear of the next cold or viral infection. For the SMA sufferer you have no independence and limited choices. Access is still an issue. Having to tell the authorities what you can't do all the time just to get support is soul destroying.

Type 2 age 18-25 years father

Cope with chronic depression, social isolation, constantly worrying for the life of my son

Type 2 age 18-25 years young adult

Physical Challenges: Due to SMA, my physical movements are very less and I am a wheelchair user. I am only able to move my finger unaided and all the rest I need support. for example: one of the major challenges is that I cannot drive my wheelchair properly as my finger keeps on slipping from the joystick. I need someone with me 24 x 7. This makes me frustrated. I am mentally capable but physically I cannot do anything. I am on very heavy medication for 24 hours. I need 8 physio session with the use of cough assist and suction machine. In spite of doing all this, I get recurrent chest infections. This medical treatment and physiotherapy takes lot of my time which otherwise I would have used for my other personal and social needs. I always live in fear of getting unwell and which may lead to hospital admissions. Due to my physical movement restrictions and dependencies on machines, I have not gone to any holidays for years now. This also affects my parents' movements as they have to look after me continuously. Emotional Challenges: I cannot do things like other people of my age as I am continuously surrounded by carers and I have no privacy. As I am always with someone or the other, I do not have freedom of expression. Practical Challenges: the focus of my life

is only keeping me well. I cannot do anything as my will. Out of 24 hours, 20 hours the focus is on health. I can't access many places even in London.

Type 2 age 26-35 years adult

Like most people with SMA I'm intelligent and keen to participate in work and with friends but staying healthy is like running the wrong way on an escalator because it's a battle that you can't win. It is emotionally and practically difficult to rely on another person for every physical need. It's exhausting to fight for every bit of funded help and awful to watch my parents to struggle to meet my needs as they are ageing.

Type 2 age 26-35 years adult

One of the challenges in terms of quality of life is the difficulty with my social care situation and having to really fight for the funding to secure the level of care that I need. This causes a lot of emotional distress for both myself and my unpaid carer and can manifest physically too as weakness and fatigue in my body. Another big challenge is having to constantly adapt as the condition deteriorates - this has a big impact on me professionally as I have to keep reassessing my capabilities and adjusting to my new needs. The biggest challenge though is probably the feelings of fear and loss that come with the condition. I am not worried about never being able to walk or having weak arms. I have accepted those things. What I really worry about is that eventually I will have no function left and will not be able to work (which I really enjoy, I work in healthcare research). I graduated from a top university and spent 7 years training to be a scientist so that I could help others and make a contribution to society, I lie awake at night worrying about how long I have left before my body becomes too weak for me to work. I also worry because I have a partner and I want to be able to enjoy our life together. I can handle the loss I've experienced so far, all I want is a chance at maintaining my current levels of function which are still good (well as good as they can be for someone with SMA!).

Type 2 age 26-35 years father

Can't have a meal without the fear of them choking Worried what will happen when I can no longer be there or able to offer support due to illness Having to go over the same questions again and again with social services and government agencies They won't get better at present so why keep asking us to prove it

Type 2 age 36 – 45 years adult

My strength is getting less and less. I used to be able to crawl and sit. Now holding my own neck up and swallowing food is becoming problematic. I rely on help to do things I want/used to be able to do easily. For example, I can't roll in bed anymore and need so much help that it disturbs my sleep. As I am over 18 the support I get is minimal (physio, neurological support) yet I am not improving. The GPs don't know what else to do to help me. My bladder is getting weaker too- emotionally this bothers me. My life is great but for the SMA. I feel like a burden and I can't stop myself getting weaker. I have been able to walk since age 11 but I could always sit unaided. I'm now struggling to sit unaided. I live in fear I will get a chest infection and die like my younger brother did (who also had type 2 SMA).

Type 2 age 46-55 years adult

I am 52 years old with Type 2 SMA. My neurologist treats me as a palliative care patient which means I am facing the end of my life. With regard to my body and my own mind I feel I am at peace with this. However, my son is 12 years old and I feel that he is still too young to lose his mother. For me this is the biggest emotional challenge. Physically my biggest challenges are about minimising my pain levels and maintaining my breathing and swallowing ability. I have been offered a Gtube so I don't have to worry about swallowing but feel at the moment that would be an intrusion too far. I also have a constant challenge to avoid and manage chest infections as I wish to avoid hospitalization at all costs. I use nocturnal BIPAP, the mask is uncomfortable but it is essential. All aspects of my daily life are affected by my SMA, it makes all the practicality of daily routines complicated and time consuming. I have a wonderful set of 3 PAs that enable me to be a mother and do all the tasks I need to do.

Type 1 / 2 age 46-55 years adult

Navigating the NHS with a severe physical disability is, frankly, untenable – having had a recent procedure in my local, main, hospital, it is only those professionals that I have managed to speak to prior the procedure who actually were able to help through many difficulties during what should have been something quite straightforward.....I actually had to discharge myself and come home in order to have all the equipment that I needed, and my PAs whom I've trained, in order to be comfortable, adequately assisted with my requirements, and frankly, not as exhausting as having to try to get hospital staff to understand I needed.....There is no adequate emotional support, there are few medical professionals who actually understand what I need – I have an amazing OT, and equally amazing wheelchair services whose head engineer begin 'gets me', and therefore is incredibly useful and lovely. I have a great GP, but if she is not there, I am stuffed..... I employ my own PAs through a grant from the CHC, and it is a nightmare finding adequate, let alone good, PAs.

Type 1 / 2 age 46-55 years adult

Pain and fatigue make most days unbearable, I have recently started to experience increasing issues with swallowing...This severely limits the options available for pain control (timed-release mechanisms, for example). The fatigue and eating has recently begun to impact upon my food intake, and I am losing weight sporadically less and less. I am going to have a PEG tube fitted soon, which is going to be very difficult to place due to a very severe scoliosis/kyphosis I am concerned that many medications that I need, for instance, esomeprazole, do not come in a form that can go through a feeding tube about completely negating the delivery mechanism..

EVERYTHING - Apart from once I have been assisted with my arms being placed in the correct position at my adapted 2 mice – this was funded by a charity, and is a bit ropey now and I can't really go back for more assistance. Then, provided my dictation is working – and this is now an issue due to enough voice consistently – and failing that, I am up to the fatigue of using an on-screen keyboard, I am mainly 'good-to-go'. However, I need more assistance with not being able to mechanically tilt my wheelchair for drinking, and find this is causing me increasingly to have swallowing issues and therefore I choked – a lot, probably quite severely at least 12/14 days. But although I can drive

wheelchair, which is another exclusion, I am becoming less able to do so outside of the property and it is virtually impossible when the weather is even slightly colder, as my hands very quickly become unable to move. Even in good weather, I am finding I'm having to stop and take breaks – for example flat and relatively warm places such as hospitals. I am in the process of going onto a mini joystick, given my positional difficulties and many other factors, this is taking a long time, and in the meantime, I need to have my exceedingly expensive, initially self-funded previous model powered wheelchair, replaced due to its age and the fact that the company is no longer functioning. I'm petrified I will be left without my wheelchair and rely heavily on the seat position changes to alleviate pain, allow for some self-feeding and access to drinking, and literally too many things to write down