CADTH

CADTH Reimbursement Review
Patient Input Submission for CADTH Reimbursement Reviews

Name of the Drug and Indication	**nusinersen (Spinraza)** ***Spinraza (nusinersen) is indicated for the treatment of 5q Spinal Muscular Atrophy (SMA)***
Name of the Patient Group	**Muscular Dystrophy Canada**
Author of the Submission	**Homira Osman**

1. About Your Patient Group

Muscular Dystrophy Canada is registered with CADTH. www.muscle.ca

Muscular Dystrophy Canada (MDC) supports people affected by hereditary and immune-mediated conditions where the primary effect is on the muscles, the neuromuscular junction and/or peripheral nerves. These include the following disorder groups:

- Primary disorders of the muscle (muscular dystrophies, hereditary or immune-mediated myopathies),
- Neuromuscular junctions disorders (hereditary or immune mediated myasthenic conditions)
- Primary disorders of the peripheral nervous system (hereditary motor and sensory neuropathies; immune-mediated neuropathies; lower motor neuron disorders)

The specific types of muscle affected, the severity, and age at which symptoms begin to show depend on factors such as the individuals' exact diagnosis. Commonly persons living with neuromuscular disorders experience some level of muscle weakness. This may affect their arms and legs, and in some disorders the muscles needed for eating, speaking, breathing, heart and eye function maybe affected as well. Some neuromuscular disorders have multisystem effects and might affect other parts of the body such as the endocrine system, cognitive function, and gastrointestinal system. For a very small subset of neuromuscular disorders, like Spinal Muscular Atrophy, life-changing treatments are now available but access is limited; the majority of neuromuscular disorders have no definitive cures.

Since 1954, Muscular Dystrophy Canada has been the leading health charity and voice of the neuromuscular community in Canada. MDC's mission is to enhance the lives of those impacted by neuromuscular disorders by continually working to provide ongoing support and resources while relentlessly searching for cures through well-funded research. MDC represents over 50,000 registered individuals including those impacted by neuromuscular disorders themselves, family members/caregivers, healthcare professionals, and researchers. MDC supports individuals impacted by neuromuscular disorders by investing in research, delivering critical programs and services, and challenging public policy. Our services and programs play a crucial role in informing and supporting members of the neuromuscular community by funding equipment and assistive technologies to improve daily life, hosting family and caregiver retreats, providing emotional and educational support, and providing access to vital resources and support systems.

Funded by Canadians from coast to coast, our investment in the research community is advancing the development of important new treatments. Our programs and services play a critical role in improving access to vital resources and support systems and ultimately, affecting quality of life. Our advocacy efforts focus on enhancing public policy at all levels of government to bring about positive change. We are currently working to bring new treatments and trials to Canada. Advances in medicine have resulted in individuals with neuromuscular disorders living longer but not necessarily living

better. As their disorder progresses and changes, so do their needs and financial strains. MDC recently completed its own health economics research study to best determine the costs patients incur during their neuromuscular journey. These "hidden costs" include all out-of-pocket expenses (e.g. genetic testing, home modifications, mobility devices), any loss of income due to loss of employment or forced early retirement, or any of the time provided by family care-givers. The results of our study reinforce the value proposition for access to life-changing treatments and supports.

Our desire is to provide support to an individual and family through all stages of disease progression by providing the tools, resources and support to live a full and rich life and at the same time, invest in research and real-world evidence generation to support public policies and influence positive change.

About SMA

Spinal Muscular Atrophy (5q SMA) is a neuromuscular disorder caused by biallelic mutations in the SMN1 gene. There are different clinical subtypes of SMA based on age of onset, function, and outcome. The most common subtype is SMA Type 1, which presents with symptoms in the first 6 months of life, and untreated results in death typically before the age of 2 (refer to: https://www.youtube.com/watch?v=EG8zMxZeOOs). SMA subtype corresponds with the number of genetic copies of a modifier gene called SMN2. Both SMN1 and SMN2 encode for SMN protein. Patients with SMA Type 1 most commonly have two copies of SMN2, while patients with Type 2 typically have 3 copies, though can have 2 or 4. Therapeutic strategies for SMA have centred on increasing the amount of SMN protein, either by acting on SMN2 (Spinraza, Risdiplam) or SMN1 (Zolgensma). Nusinersen (Spinraza), has been approved by Health Canada since July 2017 for the treatment of 5q spinal muscular atrophy. This treatment has been recommended for reimbursement by Canadian Drug Expert Committee CDEC and MDC has previously submitted patient input submissions in support of providing access and reimbursement for nusinersen. To date, most children with SMA registered with MDC are receiving nusinersen as a part of their treatment regimen; adults affected by SMA in Quebec are also receiving nusinersen as part of their treatment regimen. Since our patient input submissions in 2017/2018, emerging evidence and real-world data have expanded our knowledge on safety and efficacy of the drug in a much larger population of SMA patients than those reported in the initial studies.

We hope this submission will demonstrate the unmet need for treatment – particularly for those above the age of 25 affected by SMA – and will reinforce the importance of access to this life-changing treatment. Please note, in addition to this written submission, MDC has conducted short video-interviews with adults in Quebec who have received consistent access to Spinraza; these are available for your review and consideration.

2. Information Gathering

CADTH is interested in hearing from a wide range of patients and caregivers in this patient input submission. Describe how you gathered the perspectives: for example, by interviews, focus groups, or survey; personal experience; or a combination of these. Where possible, include **when** the data were gathered; if data were gathered **in Canada** or elsewhere; demographics of the respondents; and **how many** patients, caregivers, and individuals with experience with the drug in review contributed insights. We will use this background to better understand the context of the perspectives shared.

Muscular Dystrophy Canada has Neuromuscular Service Support Staff in all provinces across Canada. As part of the System Navigation Program, the Neuromuscular Service Support Staff provide front-line support to thousands of Canadians affected by neuromuscular disorders. The program operates on collaboration and patient engagement principles. Neuromuscular Service Support Staff work directly with patients and family members to identify non-medical needs (e.g., housing, transportation, access to equipment) and provide them access to the right resources in a personalized customized manner. Neuromuscular Service Support Staff work in partnership with patients and their families to address barriers, network and make connections with others in the community, share education materials and resources, enhance life skills and self-coping strategies, embrace inclusion and ultimately provide supports to help positively improve the overall well-being and quality of life of the patient and their family members.

The Neuromuscular Service Support Staff identified and contacted adults living with spinal muscular atrophy to participate in a healthcare experience survey (available in English and French) and semi-structured virtual (phone, Zoom) interviews.

The following submission reflects data from 60 individuals (age 18+) impacted by spinal muscular atrophy. Each respondent had a confirmed diagnosis of 5q SMA, as signed off/confirmed by a neuromuscular specialist on their registration form. The respondents included 31 males and 29 females. Respondents were between ages of 19 – 80, with responses from adults affected by SMA in Quebec (n=20), Ontario, British Columbia, Alberta, Manitoba, Saskatchewan, New Brunswick, Nova Scotia and Prince Edward Island. The responses were collected from December 2021 to January 4, 2022.

We sought the opinion on the value of having Spinraza approved for adults affected by SMA in Canada. A qualitative descriptive approach, employing the technique of constant comparison, was used to produce a thematic analysis. We have included patients' quotes to ensure their voices are captured in this report and to provide context for quantitative elements. A report capturing all patient comments is also available for review.

3. Disease Experience

CADTH involves clinical experts in every review to explain disease progression and treatment goals. Here we are interested in understanding the illness from a patient's perspective. Describe how the disease impacts patients' and caregivers' day-to-day life and quality of life. Are there any aspects of the illness that are more important to control than others?

In response to the question posed by MDC: "Can you describe how SMA impacts your day-to-day life and quality of life?" - the following 5 key themes were identified (in order of frequently reported): 1- significant impact on independence; 2- significant impact on activities of daily living; 3- negative impact on mental health and well-being; 4- negative impact on energy levels (fatigue); 5- negative impact on work participation. The below quotes from individuals affected by SMA highlight that the impact of SMA on adults is not purely physical, but that the condition impacts mental health, quality of life and the well-being of families.

Impact on Independence

*"**Unable to live day to day life independently**. Extreme muscle weakness, fatigue, pain and endless tests and doctors' appointments while trying to live like every other member of society. It is impossible to have a true quality of life without treatment, knowing any day could be your last."*

*"SMA atrophies muscles and day to day quality of life**. It atrophies independence and challenges life in every aspect.** The fact it is degenerative you never know what is going to be lost the next day."*

*"SMA has taken a lot of physical abilities away from me. **I can no longer feed myself, dress myself, brush my own teeth, or grab a simple glass of water.** I do have a good quality of life thanks to my amazing family that does not let my disability stop me from anything, but it is an exhausting disease nonetheless. It's tiring relying on other people 24/7 and going through the emotional roller-coaster of losing function (prior to treatment)."*

*"Spinal muscular atrophy (type 2) **greatly limits my independence**. It is difficult, or perhaps impossible, to point to an aspect of my life that is untouched by this disease. Feeding, transfers, personal care, and transportation are all severely different than those without this disease. My daily life is also greatly changed as I require help with most simple tasks (drinking, completing school-work, showering, toileting, dressing, etc.). The activities that I can engage in, and places I can visit, are strictly limited by the accessibility of buildings/areas (for example: the height of my wheelchair limits my ability to sit at tables of regular height, I can only sit at tables that are bar height).*

"SMA contributes to body muscle weakness and that involves being confined to a wheelchair at all times. It also means that over my lifetime my muscles will atrophy and grow weaker because I do not produce the protein for muscle development. ***I require assistance for most aspects of my day-to-day living, and that level of assistance will only continue to increase as I get older because I will get weaker. If I did not have a stable family unit and comfortable household income my quality of life would most definitely be a struggle, as the amount of health care aide I require exceeds what the government can supply for me.*** *I'm fortunate, but there are many in my position who have to live in full-time care facilities to have their basic needs met. As you can imagine, someone's quality of life in that type of situation would not be very high."*

"I ***require 24hr support of a caregiver for all aspects of my day-to-day life****. From dressing to meal prep and assistance using the washroom. SMA makes it so that every step in my day is a little more challenging to achieve a healthy quality of life."*

"It's hard to get comfortable while sleeping because I can't move around easy. I need a hospital bed to raise me up enough to stand. I need to use two canes to walk, which is increasingly becoming harder and I worry I'm about to lose my ability to do so. ***I moved back in with my parents 4 years ago after living alone for 14 years because I was getting to weak to live on my own, and have since become a lot weaker. I now require help doing daily activities that I didn't need help with even a couple months ago, like getting out of the shower or off the toilet.*** *I don't leave the house much more than once or twice a month because I get tired too easily now. Over-use of my "good" limbs that have gotten weaker causes a lot of pain, including bicep tendinitis and rotator cuff issues in my right arm, which is the only arm I can lift."*

"I ***require assistance with all aspects of my life by specially trained caregivers****. I am ventilator dependent and unable to swallow. Presently I am able to move my thumbs."*

"SMA has affected my ability to eat, walk, dress and toilet independently. ***I rely on someone else for all aspects of personal care and cannot live independently****. I have chronic pain and my mental health has been impacted due to SMA."*

"I have very little movement in my body so ***I am 100% dependent.*** *I can't feed myself or even move my arms. Swallowing is tricky so eating takes a great deal of time. I have pain if my body is not in the right position. The biggest issue is my breathing which is extremely shallow. I have stayed in my house outside of the summer season for most of my life because of risk of infection from viruses. My social life is all virtual because of this."*

"Every single moment of every single day I am affected by SMA in one way or another. ***I cannot get into bed on my own and rely on home care to come every morning to get me dressed and showered****."*

*"****I need to have a caregiver around all the time, or at least someone who is willing to do my care****. In high school I couldn't just go to a friends house for a sleepover, because I need to be repositioned at night and use my bipap. In college, I couldn't just go on a blind date on my own, because I need help with feeding / drinking. As an adult, I struggle to have autonomy in my life and career because I am relying on someone else's schedule 100% of the time."*

"I cannot drive, get to Dr appointments, get groceries, do laundry, cook myself a meal, work a normal job, clear secretions to keep myself from choking or truly have any type of independence outside my home."

"SMA has really ***affected my independence because I was able to walk and now I'm stuck in an electric wheelchair and requiring help all the time.*** *I'm too weak to drive. I don't feel comfortable anymore going outside or doing an activity by myself anymore because going to the washroom has become almost impossible. Being an electric chair causes so much problems because you can't fold up a power chair it's really bulky heavy. In addition, because there's no funding*

for manual chairs, you have to pick one. I've always wanted to slow dance with a beautiful woman standing and that will never happen."

"It has affected my ability to do virtually anything independently, other than drive my specialized wheelchair and type on my iPhone."

*"I have **no independence**, I am completely dependent on other people which restricts my ability to move out, travel, and forces me to arrange my life around other people (caretakers getting sick, needing vacation, etc)."*

"I can't do my personal care. It's becoming more difficult to go out on my own especially in winter (because of extra clothes)."

"I can't live alone anymore. I need help getting into and out of the shower. I need help getting off the toilet. I need help getting items out of the freezer or cupboards to make my own meals. I need to be driven everywhere by one of my parents, and even if I could drive on my own, it'd be too expensive to ever buy one of those vehicles. I need help going to the bathroom in public places. I need help transferring to the eye doctor or dentist chair. I need help with shopping because I can't carry too much or reach some items. I need help plugging in my wheelchair to charge it because my hands have become too weak. I need someone to do my laundry for me and make my bed and put my clothes away. Basically, shortly after turning 30, I started to get increasingly weaker and lost a lot of my abilities and nearly all of my independence. I've lost most of my abilities just in the last 4 years since Spinraza was approved by Health Canada but not accessible to type 3 adults. For example, in 2017 I was able to babysit my infant niece of my own for short periods of time, lift her on my own, get her out of her crib on my own, take her outside to play on my own, cook for her and feed her, change her diaper etc. Now, I can barely hold my new 5 month old niece on my own while sitting down without my arms giving out, and sure as heck can't lift her, change her diaper or anything of the sort."

"I cannot live independently and rely on others for assistance. I need someone to accompany me to appointments and community activities."

"I am in a motorized wheelchair. I require a caregiver to perform personal tasks. I also have specialized equipment to help me at work."

Impact on Activities of Daily Living

*"I require the use of an electric wheelchair, **I have to have attendant care for even the basic things like going to the washroom bathing changing transferring.** As I get weaker, I require more care and less I can do."*

*"It is hard for him to me to work on the computer. **Everything, toileting dressing, transfers and even eating is getting more difficult because it is harder to hold utensils.**"*

*"SMA affects everything about it. Everything is a chore for me. **I am at the point right now where I have a hard time getting out of chairs. It affects me constantly.** I am okay and can get around, but if I sit in a chair with no sides I have a hard time getting up."*

*"Having SMA my entire life and in a wheelchair since I was 15, it is hard in just about every way. **The biggest impact is the progressive nature that I cannot count on being the same person in any type of future.** My body, mobility, strength, and function in the future is unknown."*

*"I **need more and more help in doing all my daily activities.** My medical needs and appointments are consuming a lot of my time and resources. I decided to go through surgeries to manager my bladder and bowels without transferring to the toilet. The structural changes improved my life, but they certainly come with some issues."*

"I depend every day on a caregiver, my wife ***need help to use bathroom, shower, getting dress, meals.*** *I cannot walk, I have a powered wheelchair and lifts."*

*"****It limits me in most capacities through my day including waking up, getting dressed, preparing meals, caring for my son, cleaning my home, attending my appointment, basically every aspect of my life is carefully planned out and coordinated so that I'm able to continue to continue to be a healthy human.*** *My disease affects my personal finances greatly, personal relationships and most importantly do to my last bout of progression my mental health has been severely compromised due to the lack of an accessible treatment for my disease which is absolutely disheartening considerate than a 4 year battle to get access to Spinraza and I'm still unable to access it."*

"SMA impacts all areas of my day to day life. I need assistance with ***dressing, showering, transferring, grooming. I use non invasive ventilation during sleep, and a cough assist machine when sick.*** *I am unable to maintain nutrition orally because of a weakened swallow, so most of my nutrition is by g-tube."*

"SMA impacts my quality of life greatly every single day from the moment I wake up; to the moment I go to bed. I need assistance with ***transfers, lifting objects, toileting, showering, dressing, and generally doing most things****."*

Impact on Mental Health & Well-Being

"SMA imparts all aspects of my life, from the mobility in my body limiting my ability to dress, eat, bathe, or perform other essential tasks of daily living. However, further to this, it ***affects my mental health*** *as well. Knowing that my body is constantly failing me rapidly gives me anxiety around my place in society and self-worth. The knowledge that there is a drug available to help that is still out of reach because of government mandates makes me feel less than my younger counterparts."*

*"****Mental health is a huge issue*** *and I have dealt with depression for most of my life. I deal with discrimination a lot of the time and ignorance from people in the public."*

"I am a very sociable person. I find people see the chair and judge me because I am in a chair as opposed to not knowing who I am as a human being."

"Emotional is having my family watch me degrade. It is hard for them to watch this, they feel bad, they are unsure of the future. Having to tell people and share medical information because it is something that is not hidden anymore. It has to be explained anywhere I go why I can't do things."

"The ***mental toll this disease takes is just as cruel as the physical toll it takes****. Living with the knowledge that your body is wasting away to the point of death. Grieving the loss of each new ability. Grieving the loss of a future that will never be.* ***All of this grief and anger and anxiety is exasperated by the fact that a treatment exists that could save your life but the government does not think you're worth it****."*

"Loss of independence ***despair;*** *financially reliant for all housing care for all personal support."*

*"****Because of me getting weaker I've become depressed, I've been suicidal even taking my life once and being rescued by paramedics.*** *People treat you differently when you become weaker. Or people think you're the same strength when you're really not. It is so disappointing when all your friends want to go to a place and you really can't go because either you're tired or it's not accessible or your chair can't fit in in the car or going to your family members house and it's not accessible because most houses are not and you missed out on a lot."*

*"****Very embarrassing****, people laugh about the fact that I cannot get up."*

"It keeps you from going to things. It is starting to confine me more. I am becoming more wanting to stay home because it takes a lot from me. I really don't like to be ***embarrassed****. When I went for my booster, I told them I couldn't sit down and they thought I was joking. It is very limiting."*

"My constant need for help is ***emotionally trying*** *and I have a hard time doing things that others take for granted."*

*"****The need to rely on my elderly parents is emotionally taxing*** *because they have their own issues. I am less comfortable going out on my own to social gathering, and I do not want to have my caregivers with me everywhere I go. I cannot enjoy my baby niece as I would love too because my arms are getting weaker and weaker. She is my biggest joy in life but I cannot lift her to smell her and kiss her."*

"The one that immediately comes to mind is finding and maintaining a romantic relationship. It's hard for many to see past a wheelchair and obvious body deformity but that's not to say it's impossible either. I have maintained strong social and emotional connections with friends and partners, but I think I might be the exception to the rule on that one. I've had conversations with others who have SMA and they have told me they ***felt ostracized for much of their life****. Living with SMA means you have to be able to overcome* ***many social hurdles*** *before being able to form a connection with people. I was able to do this, but many are not."*

"Knowing that I'll continue to get weaker and the ***anxiety that comes with knowing that to the point that I've had to go on an SSRI medication for anxiety and depression.*** *Constantly thinking of all the things I used to be able to do and can't any more is emotionally draining.* ***I don't really have a social life or friends,*** *I rarely leave the house because it's too tiring and I have to rely on my parents to drive me because I can no longer live on my own in the city.* ***Knowing that I'm dependent on my parents and feeling like a burden at times is frustrating*** *and I wish I didn't have to put that responsibility on them. I've always felt ashamed/embarrassed about not having a job after college, which made me avoid old friends and acquaintances. I've never been on a date or had a romantic partner and I'll never have a family of my own. I worry about the future a lot and what I'll do when my parents die or can no longer take care of me."*

"Feeling ***excluded from events or social situations*** *because of a lack of accessibility."*

"Severe depression and fatigue."

"I am not always able to do activities I enjoy because of my energy and health, and the lack of financial resources (for hiring caregivers). As a result, ***I have bad mental health****."*

"Life is a roller coaster. You never know what you are going to lose next. You constantly are fighting battles of some sort or another. ***It's extremely wearing on not only myself but those around me****."*

"Growing up, seeing your friends gain more independence while you remain the same. As an adult, this is slightly less but the same thing does happen. Being left out of things because you cannot participate. ***The mental/emotional impacts of realizing that you lost the ability to do something as the condition progresses****."*

"Fatigue, inability to feed oneself, reliability on others for personal care can ***greatly hinder the ability to participate in social activities****."*

"For me, the ***isolation from in person contact is depressing****. I have to avoid crowds and I do not even leave my home for 9 months per year. I have a good virtual life but it's not the same."*

"I ***feel isolated around everyone because my life and body is so different from theirs****."*

*"I have often felt **very socially isolated.** What lockdown was like during the beginning of the pandemic that is what most winters are like for me."*

*"**The social and emotional consequences of SMA are almost more powerful to me than the physical ones**. I often struggle with feelings of being a burden, being unworthy of love, being unable to participate in society in meaningful ways, limited freedom of choice in terms of where I can live, where I can go, and who I can spend time with. As a 30-year-old, as very much affected dating, where people often see me wheelchair-first and gives me profound feelings as emptiness and loneliness."*

Impact on Energy Level (Fatigue)

*"I am very active person so **managing my energy is almost like a full-time job**. I have to choose what I do so I am not too tired or get weaker."*

*"I deal daily with shortness of breath, weakness, **fatigue** and cramps."*

*"I'm having **less and less energy**. I'm less able to participate actively in the community and enjoy my social life."*

*"Quite literally every aspect even down to the task of breathing is affected. **The tasks that the majority of people don't even think about their body doing, such as breathing, takes a lot of energy and focus for my body**."*

*"SMA makes it difficult to do tasks and **I get tired very easily**."*

*"My **energy levels are low** so I often cannot do much before needing to rest. Because of having to be full-time in a power wheelchair I have extremely sore hips and back and struggle often with pressure sores."*

*"**The fatigue limits my ability to socialize, and restricts my functional time to work and study.** Additionally, having to rely on other people 24/7 is a **crushing weight on my mental health and self-worth**, as well places a strain on these relationships."*

*"I hold myself back from activities and experiences because it will be **too much energy usage to do it**."*

Impact on Work Participation

*"I have very limited use of my four limbs. My dexterity is extremely difficult to even bend my fingers. **I used to work for the Toronto star for 25 years. When I turned 51 I became very fatigued so I had to go on long-term disability.** I am in a motorized chair and I do have PSW's come in to help in the morning and night."*

*"It has been progressive. I am a father of three and especially over the last two years, I cannot do any long distance walks (a block), I can no longer climb stairs. **I had to advise work of my condition. I had to share with everyone that in the new future I would need some assistance.** The impact is the slow degradation and loss of the ability to do things. Also, the inability to do things with my family."*

*SMA affects every aspect of my life. Living confined to a wheelchair with extremely weak muscles limits my ability to do anything normally. I rely on others 24 hours a day to help me with most aspects of my life**. I am able to continue to hold down a job and function fairly well at it with help. As my muscles weaken, I don't know how long that will continue.**"*

*"**Maintaining a career or position outside of my home is incredibly difficult**, as I would have to pay someone to be with me most of the time. So I have chosen not to pursue a typical nine to five job and take advantage of opportunities*

from my home. I was able to complete a bachelor's degree in Arts and have the option of pursuing a masters or PhD, but again this was only possible due to my generational wealth and healthy familial unit."

***"I haven't worked in 9 years.** It can be very depressing."*

*"In the past, I have had to fight my employer in order to have basic needs not **in order to keep employment**."*

*"Due to **exhaustion** and decline in my ability to type, I am on long-term disability with my employer until I retire."*

4. Experiences With Currently Available Treatments

CADTH examines the clinical benefit and cost-effectiveness of new drugs compared with currently available treatments. We can use this information to evaluate how well the drug under review might address gaps if current therapies fall short for patients and caregivers.

Describe how well patients and caregivers are managing their illnesses with currently available treatments (please specify treatments). Consider benefits seen, and side effects experienced and their management. Also consider any difficulties accessing treatment (cost, travel to clinic, time off work) and receiving treatment (swallowing pills, infusion lines).

In response to the question posed by MDC: "How are you managing SMA with currently available treatments or therapies. For each therapy what are the benefits seen, and side effects experienced? Do you have any difficulties accessing these treatments?" - the following 3 key themes emerged: no treatment experience; positive benefits of Risdiplam observed; opted for alternative ways to manage SMA.

The below quotes from individuals affected by SMA highlight the significant treatment gap for adults.

No treatment experience

"No types of medication."

"No therapies at the moment for me."

"I have not had any treatment experiences. I live in Ontario, my brother lives in Quebec and he started treatment with Spinraza a year ago because of my age. This is very frustrating. I am hearing that they are seeing improvements in my brother and the Ontario government is saying it doesn't help."

*"**I am an adult and currently no treatment is available for me. It's criminal.** Especially when most of the world covers SMA drugs for adults."*

"No experience with treatment."

"I feel as though I have not had any treatment my whole life as any procedures that I have had done are due to the progression of my disease, rather than a treatment for improvements."

"No treatment. No medication for adults like me affected by SMA."

"I've never been granted an opportunity for treatment."

"I have had no treatments."

"I haven't had any drug experiences."

"I wasn't able to access any SMA treatments."

"To date no treatment options have been available."

Positive treatment experience with Spinraza initially; switched to Risdiplam

"I have done 4 Spinraza doses and Risdiplam for 3.5 years. They both worked well. Only reason I switched was because Spinraza was not funded and I got accepted into a clinical trial for Risdisplam. I was on a trial for Cytokinetics (a muscle activator) I saw muscle improvements with this medication. I was on this study for 4 months, which was the length of the study."

"I have been fortunate enough to receive both Spinraza (7 doses, I believe) and now Risdiplam. ***On Spinraza I noticed a MASSIVE energy boost after each shot. It was truly unbelievable.*** *I went from needing 12 hours of sleep at night to 7. I was working a full time (in office) internship for school at the time and I had absolutely no issues keeping up or maintain energy. In fact, I actually had a second summer job on the weekends! During this time, my trunk and strength also significantly improved, as well as hand movements. The only downside was that the injections were painful and due to my scoliosis, it was very challenging for them to access my spinal fluid. On my last dose, the radiologist tried for about 2 hours but was unable to get to the right spot. This is when we made the decision to switch over to Risdiplam, since it's oral. On Risdiplam, I've been seeing positive improvements too. Most noticeably is my increased lung capacity; I can now nap without my bipap (which I lost the ability to do in elementary school), I cough and sneeze so much stronger, and my voice is so loud and clear. I also gained the ability to whistle! Physically, I can now wiggle my toes and I have much better neck control overall. I believe my grip in my hands is stronger too."*

"I am currently on Risdiplam/Evrysdi and have seen slightly improvements in my energy and endurance. My swallowing is slightly better as well."

"I have been taking Risdiplam for a few months. I've seen moderate improvements and had no side effects."

Opted for alternative ways to manage SMA

"Traditional Chinese medicine improved circulation. Heel cord lengthening and mobility aids have made it easier to get around."

"I took ***creatine*** *for about 5 years in early 2000's with marginal benefits."*

"Besides ***physio****, I've never been treated."*

"Currently I am not receiving any drug treatments for SMA. The only "treatment" I am receiving is the use of a BiPap at night and ***Cough Assist*** *during the day to try to maintain my lung functioning for as long as possible. These practices have so far been able to slow the loss of lung functioning, but act as more of a deterrent and less of a solution. These machines are in my own home, I am required to see a doctor/respiratory therapist once a year to maintain them."*

"As far as treatment goes, there hasn't been much. I received an operation when I was 10 to implant metal rods in my spine for support. Without this operation, my chest cavity would have declined to the point of crushing my organs I'm bending my spine. This was a major operation and a lot for a 10-year-old, I don't even think I realized the scope of what was happening to me at the time. But it worked and prolonged my life. ***The only other thing I could consider treatment is physiotherapy****, and while it helps, it's simply a means of prolonging the inevitable."*

*"To be honest, I have never really treated SMA. I never took medicine for SMA. I used to do physical exercise but it didn't show improvement and eventually became more hassle and pain then it was worth****. Swimming was good*** *but again, eventually became too difficult and/or painful."*

"Because of COVID I have not seen my specialist in some time. I find staying active to be the best thing for me.

*Unfortunately I wasn't offered any treatment, for example **physiotherapy** until I broke my femur and fibula bones in my right leg. This began a journey back to strengthening my core and hope ultimately to be able to stand and walk again. I had resigned to my diagnosis and hadn't exercised. I wish this had been offered or even suggested to me."*

5. Improved Outcomes

CADTH is interested in patients' views on what outcomes we should consider when evaluating new therapies. What improvements would patients and caregivers like to see in a new treatment that is not achieved in currently available treatments? How might daily life and quality of life for patients, caregivers, and families be different if the new treatment provided those desired improvements? What trade-offs do patients, families, and caregivers consider when choosing therapy?

Improvements that patients and caregivers would like to see in a new treatment can be categorized as those that (1) promote muscle strength (primarily in the arms and with respiratory function) and (2) slow down progression of disease (or reverse damage).

Improve muscle strength (primarily arm and respiratory)

*"**Arm strength and dexterity** is the most important thing that I'd like to see. It would give me more freedom. It wouldn't give me all the freedom I'd like, but a much better life."*

"I think that both Spinraza and risdiplam meet the goal of treatment. The only thing I would like to see as a secondary treatment would be a muscle activator to the gene therapy."

*"If I was able to get a little **bit of strength back in my arms** I can hold my baby niece and I keep playing the sport I love."*

*"A faster **reversal of lost strength / motor neurons** would of course be nice, but honestly I'm incredibly happy with what it's already given me. I never would have expected one treatment (never mind several) to come out in my lifetime."*

"To be able to walk up stairs and to get up off the floor or to garden."

*"The **fine motor skills** would be a big thing to improve his hands and neck. They are little improvements, but it would make a big difference."*

*"I would like to **get some of my strength**. I would re gain my social life. I would hope to be stronger and continue with my life without being in a facility. That would be the best. My life is slipping away from me."*

*"More **strength** in arms."*

*"More impact with respiratory **strength**."*

*"**More strength**, less pain and less stiff joints."*

"I wish there was something I was able to access that would improve my strength or maintain what I have."

*"Energy and **upper body strength** are the most important to me."*

*"I would like to see anything that can safely **increase or stabilize the muscles** regarding the lungs. Even a 10% increase in breathing would be a godsend. Secondary, **improvement in arm movement** would be amazing as well."*

Slow down progression of disease

*"Firstly **to stop the progression of the disease**. Secondly to **stabilize**. Thirdly to repair the atrophied muscles."*

"Just stabilize what I have would be a huge benefit. In addition, it would give me hope for even newer drugs that could potentially fully cure me. If I can get more energy, I could do so much more."

*"I would like to see a **stop in progression**, and any regain of function would be a big bonus. Neither of these are attainable with the current treatments that I am on. Daily life would become easier, I would be able to spend less time on current treatments as well as possibly regain some independence. The greatest difference would be made years down the line, as a new treatment may be the only thing preventing me from being bedridden and permanently trached."*

*"Improvement in breathing and **further slowing down of muscular weakness**."*

*"A complete stop and **reversal of symptoms**."*

"A drug that is able to replicate or allow my body to create the protein for muscle development would be huge. That is really the only way I can see my quality of life remaining stable and/or improving. I don't see robotics and brain bandwidth computing reaching the same level of quality of life improvement in my lifetime. Medical equipment and wheelchairs can always be improved, but that's no substitute for a declining body."

*"I would love to be able to regenerate muscles or at least **stop the progression**."*

*"I would absolutely love to see they're being a drug or treatment that **will slowly reverse the damage done**. I know that's asking so much and most likely not going to happen. But one can dream."*

"Any improvement inability would be amazing, but to maintain what I currently have would also be a win."

In response to *"**how might daily life and quality of life for patients, caregivers, and families be different if the new treatment provided those desired improvements?**"* the respondents shared:

"Especially in the winter I rarely go out. Only to do groceries with someone with me. I would like to go out more often, but I have to put on a coat and gloves etc and I need assistance to do that. With this treatment if I can get dexterity and arm strength I can do those things on my own and not rely on someone to do it for me. Also cooking, without my arm strength I shy away from it. I live by myself so I have to get everything done before the PSW leaves at 11am, but there's not a lot I can do."

*"It would give me **a huge sense of security**. Right now, I am trying to decide how to handle the next few years. Am I going to leave my job, which I love because I can't walk? For me, it **would keep me as an active participant in the workforce and able to do the things I can currently do.** If the progression doesn't stop soon it is unlikely I can come back from it."*

"Maintain and or gain independence emotional well being."

*"If treatments could reverse damage to the point where we could be independent in some or all of our daily life, that would have massive impact. **Less caregiver burnout, better mental health, more independence and autonomy** for the patient."*

*"I would **gain more independence with increased strength and reduced atrophy**, which would improve all aspects of my life, including my mental health and relationships with caretakers and family. The other people in my life would have more time for themselves."*

"It would make everything simpler. It would make everything better. Everything gets better. My whole life-social, mental to physical it would help me."
"I would hope to become more independent. My family wouldn't have to do as much for me."

"If it works, just being able to stay stable with a strength level or an ability level that I can count on would change a lot. I could make plans for the future and know that I can reasonably pull of the plan whether it is travel etc. Right now I can plan a trip in a year, but I have no idea what kind of shape I will be in."

"I will rely less on them. They will worry less about me. They will live their life more freely. ***The relationship between the family members will be more familial than caregiver-care recipient relationship. This will also grant us more dignity and independence****."*

"The level of improvement would be hard to quantify, but it would be huge. We're talking about ***needing less round-the-clock care and more independence for myself. Sure I won't be walking or achieving complete independence, but the reduction in cost to health care services would be astronomical on its own****. Possibly being able to drive myself, continue to feed myself and even prepare my own meals, being able to be left alone without worry of an emergency happening in my home.* ***The reduction in stress to my family members alongside the growth of independence for myself is immeasurable*** *if you ask me, and that's not even taking into account the reduced financial strain to the government over the course of my lifetime. I should also mention that treatment such as Spinraza is also potentially lifesaving, as I'm less likely to succumb to sickness such as pneumonia in my elder years. A longer life filled with fewer complications sounds like a pretty good boost to quality of life to me."*

"My husband wouldn't worry so much. I have a two-year-old grand daughter and there's not much can do with her. I can't lift her up because the added weight may make me fall."

"Patients would be less dependent on families and caregivers, health care, equipment, renovations. Patients and even their caregivers would have way more independence, freedom, happiness, a sense of purpose and way less stress, anxiety, depression."

"Quality of life would be drastically improved. I'd be more optimistic for the future and consider starting a family."

"I would be able to work more and have more social interactions with my friends and family."

"Any increase in independence would have tremendous emotional improvements on the patient. It would also free up time for families/caregivers, and provide an emotional boost for them as well."

"This would be a huge impact to clients and caregivers, families and other supports. This would also be a big impact on the healthcare system in a positive way as it would be Less costly (medical equipment, medical procedures, strain on the medical system for hospital stays etc.)"

"If my breathing increased enough so a common cold wouldn't be life threatening, then that would completely change my life. I wouldn't be so nervous around people and I would be able to go more places more often. I would have more energy and not need my caregivers to be so careful with my position as some positions make my breathing too difficult. If my arm movement increased enough then perhaps I could once again feed myself. That would be amazing."

In response to ***"trade-offs which patients and families consider when choosing therapy"*** the respondents shared:

Costs

"The largest barrier to treatment is the price tag, it is unreasonable to believe that any family can afford such preposterously priced drugs. Outside of this, there are no trade-offs I can think of that individuals and their families wouldn't hurry to embrace."

"As far as pain, that is fine. As far as money it is my life. The money issue isn't up to me, but as far as anything else I am not concerned."
"Costs mostly."

"Hospital admissions, financial resources, time."

"The only thing now would be cost. That's it."

"Cost is a big one. I could not access Risdiplam in Canada so I had to access a trial in the U.S which was a lot of travelling. I was the only Canadian accessing it. For receiving treatment, finding a doctor who is willing to administer the drug was big. Administering Spinraza with scoliosis and a rod sometimes required multiple appointments."

"There are no difficulties except financial. Many patients are unable to sustain permanent full time employment."

"Currently the Ontario government does not cover any treatment. I'm on ODSP and make little money. So travel costs money for the treatment itself would be all difficult."

"Cost, provincial access, travel to clinic, missed work hours, uncomfortable treatment options."

Potential Harm

"Does this treatment help my life more than it harms it? Personally, preventing muscle loss via treatment is the most important thing I could do for my health. So, in my opinion, a treatment would have to be really harming me to opt for no treatment."

"I understand that there ma be some pain involved in the surgery and in the recovery. I would be afraid if there was a chance of paralysis from the treatment. I wouldn't want to lose that."

"Clearly I don't want something worse than the benefits. If the medication is going to destroy organs or something it would have to be a substantial problem for me to reconsider treatment."

"If the side effects will be worse than any gains made."

"Nothing that negatively affects my breathing is even considered. It must not do that. If the therapy gets rid of my arm movement entirely, then also no. Those are the 2 most important things to me. It also goes without saying that negatively affecting my mental capabilities is out of the question. Also, the pain involved must be minimal."

Mode of Therapy Delivery

"I would say that the method of delivery. I prefer the oral drugs instead of the intrathecal because I can do it from home and it is less medical procedures for me. It is also cheaper."

"Ease of use. I live in a rural area which would be to get Spinraza I would probably have to travel 4 +plus hours to get it. I also have rods in my back."

"With Spinraza, it was just accessing my spinal fluid for to my spinal fusion rods and scoliosis. I have no difficulties with Risdiplam at all."

High Desire to Opt for Therapy Regardless of Trade-Offs

"As the patients age they are willing to trade almost anything to have new hope."

"Honestly I would do anything for therapy."

"It doesn't matter as long as I can do the things I want to do."

"Not many. If it will stop my body from atrophying I'll likely do it no matter the cost."

"Willing to drive a long distance. I would do whatever I could if there was something that would help me maintain what I have or stop the progression."

"Wouldn't choosing a therapy be a wonderful privilege?"

"I have never been eligible for any other treatment for SMA. So when Spinraza became available to me, I was willing to take the risks of the method of administration, and side effects of the drug if it meant I might attain some kind of improvement."

"At this point there is nothing that I would not try as long as it was reasonably safe."

"I am willing to do almost whatever it takes to get any sort of treatment."
The inconvenience of the procedure and the after effects would be part of it. I would need help with the financial part as well.

6. Experience With Drug Under Review

CADTH will carefully review the relevant scientific literature and clinical studies. We would like to hear from patients about their individual experiences with the new drug. This can help reviewers better understand how the drug under review meets the needs and preferences of patients, caregivers, and families.

How did patients have access to the drug under review (for example, clinical trials, private insurance)? Compared to any previous therapies patients have used, what were the benefits experienced? What were the disadvantages? How did the benefits and disadvantages impact the lives of patients, caregivers, and families? Consider side effects and if they were tolerated or how they were managed. Was the drug easier to use than previous therapies? If so, how? Are there subgroups of patients within this disease state for whom this drug is particularly helpful? In what ways? If applicable, please provide the sequencing of therapies that patients would have used prior to and after in relation to the new drug under review. Please also include a summary statement of the key values that are important to patients and caregivers with respect to the drug under review.

23 adults indicated they received the drug under review. The majority received the drug under review in Quebec; others received it through either federal health insurance plans or exceptional access. The majority reported positive benefits as it relates to strength, energy levels and slowing progression. The side effects or negative aspects were related to drug administration (invasive, requires multiple visits/travel).

*"We fundraised and my parents and grandparents all took out loans to pay for it out of pocket to qualify for the clinical trial. Benefits was a quick **increase in energy, stamina and strength**, but the downside was the **downward climb before the next dose**. I found I progressed back to where I was by the time I was due for another dose. I received some nerve damage from the intrathecal process. The cost of Spinraza was also a large disadvantage."*

*"I received it through OHIP. I was never on any previous treatments so I cannot compare to anything else taken previously. Although I will admit it was **a little painful,** I tolerated it no problem and I would 100% recommend it to anyone with SMA wanting to go on treatment."*

*"The only therapy I have ever used has been Spinraza. My private insurance approved Spinraza for me in October 2019. I have noticed **improvements in stamina**, I'm **able to do daily tasks with more ease and for longer amounts of time.** The only side effect I have experienced has been from the **administration of the drug** and not Spinraza itself. I have had Lumbar Puncture Headaches on a few occasions which are managed by laying flat."*

"I have access to Spinraza. I applied to the government of Saskatchewan, which is providing coverage on a case-by-case basis. I was approved, luckily 2 and 1/2 years ago. I am extremely proud to say that I am a resident of Saskatchewan."

"I currently have access as my province allows patients over 18 on a case-by-case basis. I have demonstrated measurable improvements and maintained abilities. ***No other treatment has done anything like this.*** *The disadvantage is that it is* ***invasive and painful****, but that is worth it. I also need to travel to receive treatment."*

"The only treatment I have experience with has been Spinraza. I started the drug in October 2019. I have noticed small ***improvements in my stamina****. I'm able to do things for longer and with more ease. At my last physiotherapy occupational therapy assessment I had gained 11 points!"*

"I have been on Spinraza for 2 and 1/2 years and it has completely changed my life for the good! Not only has my ***disease stopped*** *but I've also regained a little bit of what I lost many years ago. I will say that side effects are definitely spinal headaches and being off of my feet for a few days. Usually 3 days of rest in bad is how I manage the headaches. And of course with that comes the* ***sore back due to the lumbar puncture****. But I would do it every single day if I had to and if it meant that I would maintain the strength and abilities that I currently have."*

"I have been fortunate enough to be receiving Spinraza since June 2019. ***The results are undeniable. I received a significant increase in my strengths following my loading doses, and my abilities have not declined in the following years****."*

*"****Spinraza has helped me maintain my strength****."*

"I have a lot of benefits to receive Spinraza. Significant motor gains have been observed through post-treatment testing.
My dexterity has doubled.
My energy level has returned.
My neck is stronger therefore less dangerous in transport to injure me.
I regained the hope of living a normal life without degeneration, despite my diagnosis.
My morale has increased."

"After the first dose of Spinraza I had a lot of difficulty with heartache and headache and also a lot of vomiting. For the benefits I ***gained a lot of strength and autonomy****."*

"The first injections ***a lot of pain in my neck and my spine for 10 days.*** *I felt sick I could not eat and after those days I felt fine. My body was getting used to the injections and on the day of the injection I had side effects but after that I felt great.* ***It's like a battery that we charge after the treatment (we are weak before the treatment and after the treatment we regain strength)*** *I stopped in June because of my fat and I lost strength I am more weak in my arms and struggling to raise my glass."*

"The Spinraza treatment ***greatly stopped the degeneration****."*

*"****Spinraza resulted in better breathing, less burnout on his caregivers and less stress for them*** *- Generally less stressful for the family due to the stabilization of capacities and no degeneration."*

"Spinraza was causing me ***tremendous muscle cramps in my buttocks and quadriceps about 1-2 weeks after treatment****. I had* ***temporary energy surges*** *that lasted for about 1-2 months and dropped drastically before the next treatment. I* ***didn't have any permanent gains****. I felt the dose was not strong enough for an adult. Whether it was me or a toddler, we had the same dose."*

"The disadvantages are minimal. The treatment is complex. We have to go to the hospital. It's not super enjoyable, but ***the payoffs are well worth it.*** *The trips to go there are complex, I have to go to Montreal. The last two weeks there is fatigue. I had big side effects at first, but not anymore. I only have a little constipation.* ***For family and caregivers, there***

is no disadvantage, there are only advantages. *There was just too much expectation from those around me and there were a lot of improvements at the start, but afterwards there* ***was a plateau effect.*** *This may be due to the pool shutdown. I do not know. When I stopped swimming, I had been taking Spinraza for a year. At the beginning, I was more independent, so that freed my relatives a little. They trusted me more, they were less afraid that I would fall, that I would be alone."*

"The advantages: I regained some strength not in my arms and legs. With the help of physiotherapy, I regained strength in the buttocks, the abdominals which allows me to be more comfortable, more stable and to tolerate more long journeys in the car. In the neck too, I have less pain and it helps me to eat a little. I no longer have to take Voltaren and Tylenol for the pain. I have no side effects. The afternoon of my treatment I'm a little bumpy, but I don't have headaches and nothing else. The next day, I leave like a bomb. 2-3 weeks before the next treatment, I feel a drop. If it was closer I probably wouldn't feel that way. For my partner, who is also my caregiver, ***our life has changed*** *because I have more energy.* ***Life is better. It's less stressful for him too because my swallowing has improved so I'm less likely to choke. In my transfers I am stronger, there is a difference****."*

"I reacted positively to the first treatment. I saw ***a marked difference****. Eating and brushing my teeth had become more difficult before Spinraza. Since Spinraza, this is extremely rare. I really hit a peak and I had more core strength to support myself in my chair, my posture has improved, I no longer have shoulder pain. This is also my most significant advantage. I have had the treatments for 2 years now.* ***There is a small gradual decline, but I have kept several benefits.*** *I have had a little less strength and energy since the last year. It still remained at a very good level. I also have a small drop at the end of the 4 month. I still consider that I feel a benefit. I haven't had any side effects from the treatment itself, but I have a little question about asthma. I have developed asthma. Whether it is related to Spinraza or not, doctors cannot tell. It could be due to allergies. Only once the puncture was more difficult and I had a migraine the next day. For those close to me, the Spinraza treatments have no real impact on my family except for trips to the hospital for the treatments.* ***My family is really happy that the Spinraza works, that it relieves me, that I have improvements.*** *The only thing was my grandma kept telling me that I was easier to understand and spoke louder and looked really fit and energized. Another beneficial effect is that I eat faster than before which gives me time to do other things."*

7. Companion Diagnostic Test

If the drug in review has a companion diagnostic, please comment. Companion diagnostics are laboratory tests that provide information essential for the safe and effective use of particular therapeutic drugs. They work by detecting specific biomarkers that predict more favourable responses to certain drugs. In practice, companion diagnostics can identify patients who are likely to benefit or experience harms from particular therapies, or monitor clinical responses to optimally guide treatment adjustments.

What are patient and caregiver experiences with the biomarker testing (companion diagnostic) associated with regarding the drug under review?

Consider:

- Access to testing: for example, proximity to testing facility, availability of appointment.
- Testing: for example, how was the test done? Did testing delay the treatment from beginning? Were there any adverse effects associated with testing?
- Cost of testing: Who paid for testing? If the cost was out of pocket, what was the impact of having to pay? Were there travel costs involved?
- How patients and caregivers feel about testing: for example, understanding why the test happened, coping with anxiety while waiting for the test result, uncertainty about making a decision given the test result.

100% reported that they did have diagnostic testing completed with a muscle biopsy and/or blood test. The majority (67%) found it to be a relatively easy process, but a timely process riddled with misdiagnoses. Below are quotes that further highlight the experiences of patients and caregivers with the testing:

"It took 8 months from me starting to show symptoms to the time of diagnosis and 5 different doctors who thought I was a lazy baby. I was diagnosed through muscle biopsy, which was the only option at the time. The first biopsy was inconclusive and so they did a second one. The tests were covered because I was symptomatic and the tests were ordered by a neurologist."

"Could not get an answer in Calgary. Needed to travel to the US for genetic testing. Was told at the time would be eligible for Spinraza shot and have been waiting for over two years without a shot or any treatment. Paid $7,000 out of pocket. The testing very painful, testing was done 3 times in Calgary - went to Mayo clinic and paid $7,000 had to travel by train - 3 days."

"My genetic testing has since come back inconclusive, and they are wondering if I have a very rare and undiagnosed type. The tests that I have had done however were paid for by health care and a recent test was covered by a company that was offering testing for a certain type of MD."

"I was misdiagnosed for the first 12 years of my life. They thought it was nerve damage during pregnancy or birth. It wasn't until my dad did a bunch of research and figured out it was SMA. I don't remember much but I did get tested in Vancouver after my dad found SMA. All I remember is that they gave me a sedative because it would be painful. I just have flashes of them poking me with something. I believe that my parents were also tested for each having the recessive gene. We were nearby so no travel costs. I don't know who paid for it. I don't believe treatment changed after knowing the disability."

"I was diagnosed through muscle biopsies in the 80s and 90s, which was very painful."

"I was diagnosed with a muscle biopsy as a child."

"I was diagnosed at age 2, via a muscle biopsy."

"I was diagnosed at the age of four and I am 33 now, at first the doctor thought it was just bad parenting because I was walking on my tippy toes, it took the luck of a neurologist to be on duty that day and saw me walking and thought that I had muscular dystrophy. Some tests were done everything was covered and it was confirmed that I had spinal muscular atrophy."

"I was 20 when I was diagnosed. My mother had been taking me to the doctor since I was 2. They kept saying I would grow out of it. I had a muscle biopsy, nerve tests, a few neurologists to get to one that diagnosed me. No out of pocket tests."

"I was diagnosed in the early 70s and as such I've never really had genetic testing so far. I am looking to have this done I was initially diagnosed in Quebec. There was not a lot of experience around it. I was just told this is what it is and there's nothing you can do about it. I didn't have a document that said I had SMA, just a verbal diagnosis at first. When I moved to Ontario they did genetic testing and gave me an official diagnosis. Genetic testing was no problem."
]
"I do know that SMA was not very common and there was not a lot of information provided to my parents at the time of diagnosis. From my understanding, they had to travel from Saskatoon to Calgary for testing. I do not know if there were any adverse effects associated with the testing. They did have to pay out of pocket though to get the testing done, meaning travel to another province. I was diagnosed through a muscle biopsy when I was approximately 3 years old."
"I was born in Canada, but my family moved to New York when I was young. When I was 2-3 the family doctor did some testing and discovered I had Muscular Dystrophy and nothing after that until 14. Then they did the Muscle Biopsy and they told me I would live until the age of 30. When I was 20 I moved to Ottawa and the Civic Hospital did the EKG and the test where they did the EMG test. That is when they indicated I had SMA type II. Since then it is just my family doctor. I have been to Sunnybrook in Toronto and the doctor there I communicate with once a year and they monitor at a distance."

"My parents noticed that I wasn't moving and crawling normally as a baby. So when they took me to the doctor at the age of two they got a muscle biopsy. From what I understand it was fairly straightforward and easy with no adverse side effects. I think most of the test was covered by the government."

"I was 10. It was paid by OHIP. It was a blood test. I was tested because my brother is type 2 and the doc was curious because I walked on my toes. We drove 2 hours and spent the entire day at the hospital for the diagnosis."

"I was diagnosed 33 years ago at the age of 18 months. From what I understand, I was admitted to a children's hospital for testing where they did blood tests and a muscle biopsy. I don't believe any of this was out of pocket. There was no treatment available at that time."

"Muscle biopsy at 10 months old. To my knowledge, this test was covered."

"I had to have my diagnosis confirmed by blood test before I could begin treatment. This was done at my local hospital. I did not have any out-of-pocket expenses."

8. Anything Else?

Is there anything else specifically related to this drug review that CADTH reviewers or the expert committee should know?

"I am a person who wants to remain mobile as long as possible. I don't want to burden the system anymore than I have to. It seems from personal correspondence that Spinraza for adults has either slowed /stopped progression of SMA and in some cases shown some improvement. I don't know what else I can do as an individual to get access to Spinraza , but the roll out seems to be inconsistent and even discriminatory in some provinces."

"It's imperative that you make this recommendation because without it, provincial governments will use it as an excuse not to cover this wonderful drug that will stabilize and in some cases even improve persons with SMA. Importance of stabilization is massive for a person with a SMA. Like myself, I find myself getting weaker year-by-year. Key important things like dressing, bathing, going to the washroom Independently, playing hockey with my friends, being able to work full time, standing, walking and being able to hold a baby in my arms etc. Now these abilities are gone or fading away. I find my life becoming harder and harder to function. By stabilizing my abilities now will buy me time for future cures and better planning for my health going forward. The fear of me getting weaker will be gone. Knowing that I will be able to have the same Energy levels throughout the day is life changing! The fear of becoming so weak and becoming a bigger burden to my family, to my wife and my caregivers haunts me daily."

"If you deny coverage for the drug you'll be causing so much harm to all adult SMA patients. It is a fundamental right in Canada by law to be able to be treated from an illness without discrimination. The province of Quebec understood this and made coverage for this drug available for everyone. Please follow suit."

"Please don't sentence me to a death sentence. I need this drug to live an healthier and productive life."

"I missed out on childhood because of this rare genetic disorder and know that adulthood will be better with this life-changing treatment. We are hoping that CADTH will expand the coverage of Spinraza to include Type 2 and 3 adult patients. I do not want to lose his basic motor skills of eating, breathing, and walking. My health and future, like many other adults with SMA, depend on this treatment. We are witnessing patients in other provinces who have access to this treatment which is improving their quality of life while theirs continue to decline. As Canadians, all patients deserve to have equal access to treatment that will improve their lives."

"People with SMA are fighting for their life everyday and shouldn't have to fight this hard for an accessible treatment that is proven to work."

"I want treatment immediately, I am desperate and the coverage is very expensive to and to have funding coverage."

*"If this drug would work to help maintain or improve muscle strength everyone should have access!
Progression is going on a faster pace. Delay means more loses: physically, emotionally, socially and financially. Eventually I'll need more of the government services and healthcare system which is going to be costly too. I feel dehumanized by having the treatment out there, but not allowing me to access it although I need it badly. I've been waiting since I was 2 years old when I was first diagnosed."*

"This drug could potentially be the difference between me being able to hold my newborn baby and not being able to. This could potentially be the difference between me being able to feed myself in the future or not being able to. Do the right thing. Do it for all adults living with SMA.'

"Both Spinraza and Risdiplam have changed my life in unimaginable ways. I went from living my life in a steady (and fast) decline to a steady incline. I'm happy, I'm stronger, and most of all I'm so much healthier. I hope that every one in Canada gets access to this treatment, regardless of age, type of SMA, or geographic location."

"My entire life I have had this. I have seen countless neurologists and doctors who say there's nothing you can do. Suddenly they say, there's something you can do, but you can't afford it. That is the single most frustrating thing I have ever experienced. It is inaccessible and the government doesn't want to pitch in and its cost prohibitive. This is time sensitive. I feel myself deteriorating, any type of messing around on their part, for me that is the difference between keeping what I have and ending up with a lot less than I have."

"This isn't about being able to walk. This is a revolution in my quality of life that might as well be life-saving. I completely understand why those just recently being born with SMA have been favored to receive drugs like Spinraza. But people need to realize that it's just as life-changing for us adults too. Put me in front of anyone and I'd be happy to explain to them what this means for people like me."

"Access to this drug, regardless of age or type is essential to the SMA community. I am 34 and have had improvements and stability since starting treatment when I would surely have been experiencing a steep decline."

"My life is in your hands regarding treatment."

Appendix: Patient Group Conflict of Interest Declaration

To maintain the objectivity and credibility of the CADTH reimbursement review process, all participants in the drug review processes must disclose any real, potential, or perceived conflicts of interest. This Patient Group Conflict of Interest Declaration is required for participation. Declarations made do not negate or preclude the use of the patient group input. CADTH may contact your group with further questions, as needed.

1. Did you receive help from outside your patient group to complete this submission? If yes, please detail the help and who provided it.

   None.

2. Did you receive help from outside your patient group to collect or analyze data used in this submission? If yes, please detail the help and who provided it.

   None.

3. List any companies or organizations that have provided your group with financial payment over the past 2 years AND who may have direct or indirect interest in the drug under review.

Table 1: Financial Disclosures

Check Appropriate Dollar Range With an X. Add additional rows if necessary.

Company	$0 to 5,000	$5,001 to 10,000	$10,001 to 50,000	In Excess of $50,000
Biogen Canada				X (all funds were for educational initiatives (e.g., webinars, patient conferences) or for community engagement events; no funds received were directly or indirectly related to drug under review)

I hereby certify that I have the authority to disclose all relevant information with respect to any matter involving this patient group with a company, organization, or entity that may place this patient group in a real, potential, or perceived conflict of interest situation.

Name: Homira Osman, PhD
Position: Vice-President, Research & Public Policy
Patient Group: Muscular Dystrophy Canada
Date: **January 4, 2022**