Understanding caregiver experiences with disease-modifying therapies for spinal muscular atrophy: a qualitative study

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ABSTRACT

Objective Spinal muscular atrophy (SMA) is a neuromuscular disorder that manifests with motor deterioration and respiratory complications. The paradigm of care is shifting as disease-modifying therapies including nusinersen, onasemnogene abeparvovec and risdiplam alter the disease trajectory of SMA. The objective of this study was to explore caregivers' experiences with disease-modifying therapies for SMA.

Design Qualitative study including semistructured interviews with caregivers of children with SMA who received disease-modifying therapies. Interviews were audio recorded, transcribed verbatim, coded and analysed using content analysis.

Setting The Hospital for Sick Children (Toronto, Canada).

Results Fifteen family caregivers of children with SMA type 1 (n=5), type 2 (n=5) and type 3 (n=5) participated. There were two emerging themes and several subthemes (in parentheses): (1) inequities in access to disease-modifying therapies (variable regulatory approvals, prohibitively expensive therapies and insufficient infrastructure) and (2) patient and family experience with disease-modifying therapies (decision making, hope, fear and uncertainty).

Conclusion The caregiver experience with SMA has been transformed by the advent of disease-modifying therapies. Consistent and predictable access to disease-modifying therapies is a major concern for caregivers of children with SMA but is influenced by regulatory approvals, funding and eligibility criteria that are heterogenous across jurisdictions. Many caregivers described going to great lengths to access therapies, highlighting issues related to justice, such as equity and access. This diverse population reflects contemporary patients and families with SMA; their broad experiences may inform the healthcare delivery of other emerging orphan drugs.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Spinal muscular atrophy (SMA) is a neuromuscular disorder that manifests with motor deterioration and respiratory complications. The paradigm of care is shifting as disease-modifying therapies including nusinersen, onasemnogene abeparvovec and risdiplam alter the disease trajectory of SMA.

WHAT THIS STUDY ADDS

⇒ Consistent and predictable access to disease-modifying therapies is a major concern for caregivers of children with SMA but is influenced by regulatory approvals, funding and eligibility criteria that are heterogenous across jurisdictions.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Our results highlight the need for the effective and equitable delivery of costly, life-sustaining therapies.

INTRODUCTION

Spinal muscular atrophy (SMA) is a progressive motor neuron disorder. While there is a spectrum of disease severity, the most common and severe form (SMA type 1) is characterised by symptom onset in infancy with rapid motor decline, chronic respiratory failure and death prior to 2 years of age.[1–3]

Historically, the primary treatments for SMA were symptomatic support with respiratory therapies and enteral tube feeds to decrease pulmonary exacerbations and prolong life.[4] However, care has been revolutionised by the approval of disease-modifying therapies and the implementation of SMA in newborn screening. This has resulted in major motor and survival benefits.[5–7] In the past 7 years, disease-modifying therapies including nusinersen (Spinraza; Biogen, USA), onasemnogene abeparvovec (Zolgensma; Novartis, Switzerland) and risdiplam (Evrysdi; Roche, Switzerland) have been implemented in many countries worldwide.

In the publicly funded Canadian healthcare system, disease-modifying therapies have provincially established reimbursement criteria that enhance therapy accessibility. However, the early disease-modifying therapy era was characterised by heterogeneous experiences obtaining therapies due to the diverse availability of screening programmes locally and abroad, varying therapy eligibility criteria and prohibitively expensive therapies. Accessing therapies during the early disease-modifying therapy era was a challenge for many family caregivers.

This rapidly evolving new paradigm of care has redefined the patient and family caregiver

experience with SMA. To date, there have been four qualitative studies that have explored experiences with navigating the healthcare system to access nusinersen therapy,[8–11] but none have evaluated access to onasemnogene abeparvovec and risdiplam. A more comprehensive understanding of the caregiver and patient experience with SMA in the early stages of the disease-modifying therapy era is required to inform effective healthcare delivery and navigation. Therefore, our primary aim was to explore the patient and family experience accessing and receiving disease-modifying therapies for SMA during a unique window of opportunity in the early disease-modifying therapy era.

METHODS

A qualitative study with semistructured interviews was conducted. The study was conducted and reported in accordance with the Consolidated Criteria for Reporting Qualitative Studies.[12]

Setting and sampling

A purposive sample of caregivers to children with diverse phenotypes of SMA and ages were recruited from The Hospital for Sick Children SMA clinic to ensure a wide range of perspectives. The SMA clinic provides multidisciplinary services including neuromuscular and home mechanical ventilation care to individuals aged 0–18 years old across Ontario, Canada. The SMA clinic follows approximately 50 children, all of whom have received one or more disease-modifying therapies. Of these children, 31 are prescribed home mechanical ventilation. Currently, disease-modifying therapies are offered to all patients who meet regional reimbursement criteria, and the type of therapy is chosen in a shared-decision-making framework. Many of the children included in this study accessed disease-modifying therapies through alternative processes prior to the establishment of regional reimbursement criteria.

Subjects were identified by a member of the circle of care and recruited by the research project manager. The guardian who identified as being the person most responsible for providing and/or coordinating care for their child with SMA receiving disease-modifying therapy was invited to participate. Recruitment ended when data saturation was reached. Inclusion criteria for patients included having a diagnosis of SMA, aged 18 years or less, and having previously received or currently receiving nusinersen, onasemnogene abeparvovec and/or risdiplam.

Data collection

Semistructured interviews were conducted virtually in the participants' home using video conferencing from January to June 2022. The interview guide was developed iteratively by the research team after a review of relevant literature and consultation with content experts. It was pilot tested in five participants and involved a general discussion about experiences with disease-modifying therapies, healthcare delivery, caring for a child with SMA and social determinants of health (online supplemental eMethods 1). The interviews were conducted by a female research project manager with qualitative research experience (MA). The interviewer was not known to the participants. An interpreter was available as needed. Demographic and clinical history data were collected from the electronic medical record and during the interviews.

Data analysis

Interviews were audio recorded, transcribed verbatim and manually deidentified. The data analysis process was an inductive, four-step, content analysis approach.[13] First, five members of the research team (RA, DD, MM, SK and LX) independently conducted an immersive reading of interview transcripts to identify recurring codes. Second, emerging codes from different participant accounts were compared. Third, the research team was divided into two groups (RA, SK and LX, and DD and MM) and discussed and agreed on the codes that had emerged. Similar codes were combined, and others were reworked to form new codes. Fourth, members of the research team (RA, DD, MM, SK and LX) met twice more to reach a consensus. NVivo (QSR International, USA) was used to support the analysis. Rigour was established through prolonged engagement and peer debriefing. The participants were invited to provide feedback on the findings. This article focuses on experiences at the level of the healthcare system, whereas the analysis at the level of the individual patient and caregiver will be reported elsewhere.

Table 1 Caregiver demographic characteristics

	n (%)
Sex	
Female	11 (73)
Male	4 (27)
Relationship to child	
Mother	11 (73)
Father	4 (27)
Age range (years)	
20–29	3 (20)
30–39	8 (53)
40–49	4 (27)
Marital status	
Married or living with partner	14 (93)
Not specified	1 (7)
Education	
Some elementary school	1 (7)
Completed secondary school	1 (7)
Some postsecondary school	2 (13)
Received university or college degree	11 (73)
Employment status	
Employed full-time, working outside the home	4 (27)
Employed full-time, working from home	1 (7)
Employed part-time, working outside the home	1 (7)
Unemployed	5 (33)
Caregiver full-time	3 (17)
Not specified	1 (7)
Household income range ($)	
Less than 5000	1 (7)
5000–19 999	1 (7)
20 000–49 999	2 (14)
50 000–79 999	4 (27)
80 000 or more	6 (40)
Not specified	1 (7)

RESULTS

Eighteen family caregivers were approached for the study and 15 participated. The most common reason for non-participation was lack of availability. See tables 1 and 2 for the demographics of caregivers and their children, respectively. The mean duration of the interviews was 60 min (range 40–158 min). Fourteen interviews were completed in one session, and one interview was completed over two sessions. An interpreter was used for two interviews of non-English-speaking patients.

Table 2 Characteristics of children with SMA

	n (%)
Sex	
Female	5 (33)
Male	10 (67)
Age at interview (years)	
0–1	2 (13)
2–3	4 (27)
4–6	1 (7)
7–10	5 (33)
11+	3 (20)
SMA type	
1	5 (33)
2	5 (33)
3	5 (33)
Nusinersen	
Yes	14 (93)
No	1 (7)
Onasemnogene abeparvovec	
Yes	4 (27)
No	11 (73)
Risdiplam	
Yes	2 (13)
No	13 (87)
Distance from hospital (km)	
0–25	5 (33)
26–50	8 (53)
51+	2 (13)
Ambulatory	
Yes	5 (33)
No	10 (67)
Ventilation	
Noninvasive ventilation	3 (20)
Invasive ventilation via tracheostomy	2 (13)
None	10 (67)
Mechanical insufflation-exsufflation	
Yes	6 (40)
No	9 (60)
Enteral feed	
Gastrostomy tube	4 (27)
Gastrojejunostomy tube	2 (13)
None	9 (60)

SMA, spinal muscular atrophy.;

Two overarching themes emerged from the interviews: (1) inequities in access to disease-modifying therapies and (2) patient and family experience with disease-modifying therapies (see figure 1 and table 3).

Inequities in access to disease-modifying therapies

Caregivers identified multiple barriers to accessing disease-modifying therapies including (1) variable regulatory approvals across jurisdictions, (2) prohibitively expensive therapies and (3) insufficient infrastructure to support the rapid delivery of therapeutics.

Variable regulatory approvals across jurisdictions

The differential regulatory approval of disease-modifying therapies across the globe led some families to relocate to access life-saving therapies for their children with SMA. Several caregivers moved across continents to Canada to access multidisciplinary care for their child that was not available in their countries of origin, including the Ukraine, the Dominican Republic, Pakistan and China.

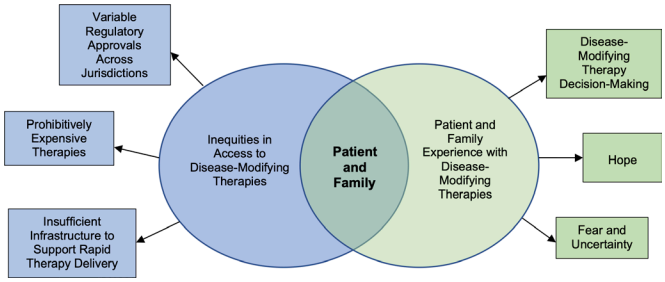

Figure 1 Conceptual model for the patient and family experience in the era of disease-modifying therapies.

Unfortunately, some caregivers were unsuccessful in obtaining sustained disease-modifying therapies after relocating to Canada. One child was initiated on nusinersen therapy but was unable to continue due to ineffective treatment response as well as the complexity of intrathecal therapy administration in the context of severe scoliosis and inability to tolerate an intrathecal port. Another family hoped to receive onasemnogene abeparvovec for their infant with SMA type 1. However, the child did not qualify for onasemnogene abeparvovec according to local eligibility criteria and was initiated on nusinersen therapy instead. The caregiver described their devastation with the arduous process of repeatedly applying for onasemnogene abeparvovec only to encounter numerous rejections (‘tell me no, don't give me that hope’ (SMA 4)).

In contrast, one patient and his family relocated to the USA to access onasemnogene abeparvovec prior to its approval in Canada, and they were successful in their endeavour.

Prohibitively expensive therapies

Access to disease-modifying therapies depends not only on regulatory approval but also on successful establishment of regional reimbursement criteria after an intense review and negotiation process. Access to costly disease-modifying therapies prior to the establishment of regional reimbursement criteria was dependent on obtaining financial coverage. Family caregivers actively sought opportunities to access disease-modifying therapies through enrolment in clinical trials, participation in therapy lotteries, fundraising and applying for compassionate use through drug companies. Many caregivers described that this process was facilitated by peer and community support, a key resource for healthcare navigation. Nevertheless, this process was extremely distressing for family caregivers who often faced multiple barriers in their attempts to access life-sustaining therapies. Despite the barriers to accessing disease-modifying therapies for their children, caregivers were highly motivated and willing to prioritise drug access at all costs.

Therapy accessibility was greatly enhanced following the establishment of regional reimbursement criteria. However, restrictive access criteria excluded some children from specific therapies and were a source of frustration for many family caregivers.

Insufficient infrastructure to support rapid therapy delivery

The urgency in diagnosing and treating SMA was highlighted by multiple family caregivers because the early delivery of

Table 3 Illustrative quotations

Inequities in access to disease-modifying therapies	
Variable regulatory approvals across jurisdictions	The care that she's receiving is very different than the care she was receiving back home. Because back home, I would never even think that there would be a respirology complex care team. (SMA 3)
	My husband was working for a company and they were opening a new store in the US. And we kind of did our research and one of the insurance companies… that was approving [onasemnogene abeparvovec] for their clients happened to be the same company that insured his company… So we applied for the transfer, and he applied for the position and got approved for it. And we went there, just like, took a chance, basically, we weren't sure if insurance would cover it, but we just, we thought we would go and try to fight for it… This is crazy, you know, but we were desperate. (SMA 15)
Prohibitively expensive therapies	We tried to advocate ourselves by fundraising, everything for [onasemnogene abeparvovec), but we received the call that they were going to choose her for the lottery to receive it that way. (SMA 13)
	Like I've said, I will do anything to get the treatment and said if I had to pay for something, I would have paid for it. Like I would even mortgage my house if I had to. (SMA 8)
Insufficient infrastructure to support rapid therapy delivery	We know that literally every moment counts. If she were to have received [disease-modifying therapy] at birth, or had the newborn screening, like it would have made her life completely different. (SMA 13)
	If a child were to come in that was in a car accident, and they could possibly lose the ability to walk if they didn't get treatment right away, they would rush them [into surgery] and make it happen. But for [my child], they just kind of like, pushed him to the side and was like "oh, he has to wait at the bottom of the list". There wasn't any urgency towards it. (SMA 15)
Patient and family experience with disease-modifying therapies	
Disease-modifying therapy decision making	We just right away knew that we were going to give it to her, like we didn't think twice about [nusinersen] because it was going to save her life. (SMA 4)
	We could go to Boston and be part of a clinical trial. Or we could stay here and get [nusinersen]… Our biggest [reason for not choosing the trial] was not knowing what his life would be like or life expectancy, we wanted to keep him as close to family, and here as we could. (SMA 5)
Hope	I don't want to be in ICU anymore. So the care goal now for this year is one year without admission. (SMA 4)
	I would love to see her walk, that would be a dream come true for me. But I also know and don't forget the reality of her condition. And all I can hope for is that her condition does not continue to deteriorate to the extent that I can't enjoy her milestones, that I can't enjoy my moments with her. (SMA 3)
Fear and uncertainty	We don't know the future side effects of it. But honestly, I think that's the last thing we think about because we know what she has and we know what could have killed her by maybe a few months after her diagnosis. (SMA 13)
	So how long does [onasemnogene abeparvovec] last? Nobody knows. So every day I wake up, every day I wanna make sure he is still healthy. And that's the feeling of fear because, maybe one day he's gonna wake up and is not going to be able to move his arm or leg or something. (SMA 10)
	What happens if the government suddenly just says your son's not eligible for it anymore? (SMA 15)
	This medicine is, the price is pretty high. And I don't know [how] the government can support this medicine… You know, maybe he got [to be] like, 20 to 22 years old, maybe he's getting older…So yeah, we'd worry about that. (SMA 6)
	I kind of questioned why we didn't get [noninvasive ventilation] sooner just for the fact of making her tolerate it when she was around six months. (SMA 13)

disease-modifying therapies is expected to result in better disease outcomes ('time is really crucial' (SMA 15)).

Systemic inefficiencies including delayed diagnosis, setbacks in obtaining drug and financial coverage approvals, and restricted health system capacity were all highlighted as barriers that delayed administration of disease-modifying therapies. One family caregiver compared the urgency assigned to a child with a limb-threatening trauma requiring surgery with the urgency of a child with SMA requiring nusinersen therapy.

Caregivers of children who received disease-modifying therapies in the presymptomatic stage of disease, enabled by newborn screening of SMA, reflected on this privilege ('it is kind of a blessing' (SMA 1)) while simultaneously expressing grief regarding the diagnosis.

Patient and family experience with disease-modifying therapies

Although the advent of therapies represented an era of hope for most caregivers, many caregivers also acknowledged the burden associated with therapies and the absence of long-term efficacy and safety data.

Disease-modifying therapy decision making

While many family caregivers discussed their desperation to access disease-modifying therapies, few reflected on the alternative of foregoing therapies. The decision to pursue disease-modifying therapies was often not viewed as a 'free choice' because the alternative scenario of severe disability or death was unimaginable.

Most family caregivers were not afforded a choice of which disease-modifying therapy to initiate due to limited therapy availability, funding and eligibility at the time of therapy initiation. For one family caregiver choosing between entering a clinical trial for onasemnogene abeparvovec abroad versus initiating nusinersen therapy locally, the decision was largely influenced by the best available evidence at the time balanced with quality of life.

Hope

Disease-modifying therapies embodied hope for many family caregivers who expressed belief that novel therapeutics would improve their child's motor development and survival ('There was no hope. But now there's hope for people' (SMA 8)). On a systemic level, some caregivers also shared their hope that these therapies would pave the way for future therapies with even greater effectiveness and availability.

Caregiver hopes and expectations of therapy outcomes were varied by their previous experiences with SMA. Caregivers of children with advanced symptoms of SMA valued clinical stability, longevity and small motor improvements or 'little inch stones' (SMA 13). In contrast, caregivers of children who received therapy while presymptomatic or minimally symptomatic expressed hopes for normalcy ('we want our son to be a normal kid. Even normal or independent' (SMA2)).

Caregivers were able to simultaneously hold onto dreams for their children while also acknowledging their pragmatic hopes with disease-modifying therapies. The caregiver for a child with SMA type 1 dependent on ventilation through a tracheostomy described dreaming of her child learning to walk while also acknowledging her primary hope for medical stability.

Fear and uncertainty

> We are lucky and unlucky. We are lucky there is a treatment. Unlucky because we are in the first line. (SMA 4)

While caregivers hoped for the best with disease-modifying therapies, there was also general acknowledgement that long-term safety and efficacy data are not available for these therapies. The lack of long-term efficacy data left one caregiver in constant fear that their child may experience a decline in health despite receiving disease-modifying therapies following newborn screening while presymptomatic.

In addition to uncertainty regarding the effectiveness of novel therapeutics, some caregivers also expressed concern regarding future eligibility and funding for medications. The expectation of life prolongation with disease-modifying therapies introduces new uncertainties regarding the transition to adulthood and ongoing provision of therapies. There were also family caregivers who reflected on the medical community's uncertainty regarding the candidacy and timing of disease-modifying therapy administration as well as initiation of supportive therapies in this contemporary landscape of care.

DISCUSSION

To our knowledge, this is the first qualitative study to explore the unique and complex experience of caregivers navigating a publicly funded healthcare system to access SMA disease-modifying therapies. Gaining access to these therapies was the goal for many family caregivers, despite the absence of long-term efficacy and safety data. Most viewed the decision to initiate such therapies as a non-choice, indicating that families would accept just about any risk if it meant the possibility of hope that their child would live. Indeed, the most prominent concern for caregivers was consistent and predictable access to disease-modifying therapies in the face of a life-limiting illness.

The willingness of family caregivers to sacrifice for the well-being of their children was unwavering despite financial, social, psychological and physical challenges. In the most extreme cases, family caregivers gave up their livelihoods by relocating to different countries to access active interventions for their children with SMA. The results of these extraordinary efforts were not always positive; some families were successful, whereas others were not due to restrictive eligibility criteria for available disease-modifying therapies. These efforts highlight issues of equity at a global level. Currently, approximately 50 out of 195 countries worldwide have regulatory approval for one of the SMA disease-modifying therapies,[14] while 13 countries have newborn screening for SMA, with many more implementing pilot programmes.[15–17]

Early experiences of accessing disease-modifying therapies have been challenging for individuals with SMA and their caregivers worldwide.[8–11] However, this is not unique to SMA. The era of precision medicine is on us, and new, costly therapies for many other diseases including cystic fibrosis and Duchenne muscular dystrophy are emerging.[18 19] Resource allocation based on an equitable, transparent process ensures procedural justice and is an important consideration in a publicly funded healthcare system such as in Canada.[20] The caregiver experiences gleaned from our study highlight the need for effective and equitable delivery of orphan drugs to allow timely access to emerging, costly, life-sustaining therapies.[21–23]

A strength of our qualitative study is the inclusion of a diverse caregiver and patient population in terms of socioeconomic status and languages spoken. We used interpreters to ensure inclusion of non-English-speaking patient populations that are historically excluded from studies. Similar to previous studies on the experience of caregivers of children with rare diseases,[8 24] this diverse patient voice highlights issues of justice, such as equity and access. Opportunities of early access to disease-modifying therapies are unequally distributed due to differential access to peer and community support for healthcare navigation, placing disadvantaged populations at a further disadvantage. Special consideration of this population is required when creating orphan drug frameworks.

Our cohort reflects a highly selected group of family caregivers who chose to initiate disease-modifying therapies for their children. We did not focus on decision making regarding the initiation of disease-modifying therapies; therefore, further exploration of this topic is needed to elucidate the contemporary decision-making process for families.

In summary, the rapidly evolving landscape of care for individuals with SMA has altered the patient and caregiver experience. Re-evaluation of current orphan drug frameworks are necessary to ensure equitable, timely access to safe and effective life-sustaining therapies. Ongoing partnerships with patients and family caregivers will ensure that multiple stakeholder perspectives are used to inform healthcare system changes.

Contributors RA conceptualised and designed the study, analysed the data, and critically reviewed and revised the manuscript. RA is the guarantor and accepts full responsibility for the finished work and the conduct of the study, had access to the data, and controlled the decision to publish. LX conceptualised and designed the study, analysed the data, drafted the initial manuscript, and critically reviewed and revised the manuscript. SK analysed the data, drafted the initial manuscript, and critically reviewed and revised the manuscript. DD and MM analysed the data and revised the manuscript. MA collected the data and critically reviewed and revised the manuscript. HG, JC, EN, EL and LW critically reviewed and revised the manuscript. All authors approved the final manuscript as submitted and agreed to be accountable for all aspects of the work.

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Patient consent for publication Not applicable.

Ethics approval This study involves human participants, and research ethics board approval was obtained from The Hospital for Sick Children (1000077616). The participants gave informed consent to participate in the study before taking part. Written electronic consent was obtained from caregivers

Provenance and peer review Not commissioned; externally peer reviewed.

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REFERENCES

1 Kolb SJ, Coffey CS, Yankey JW, *et al*. Natural history of infantile-onset spinal muscular atrophy. *Ann Neurol* 2017;82:883–91.
2 Gregoretti C, Ottonello G, Chiarini Testa MB, *et al*. Survival of patients with spinal muscular atrophy type 1. *Pediatrics* 2013;131:e1509–14.
3 Finkel RS, McDermott MP, Kaufmann P, *et al*. Observational study of spinal muscular atrophy type I and implications for clinical trials. *Neurology* 2014;83:810–7.

4 Oskoui M, Levy G, Garland CJ, *et al*. The changing natural history of spinal muscular atrophy type 1. *Neurology* 2007;69:1931–6.
5 Mendell JR, Al-Zaidy S, Shell R, *et al*. Single-dose gene-replacement therapy for spinal muscular atrophy. *N Engl J Med* 2017;377:1713–22.
6 Finkel RS, Mercuri E, Darras BT, *et al*. Nusinersen versus sham control in infantile-onset spinal muscular atrophy. *N Engl J Med* 2017;377:1723–32.
7 Darras BT, Masson R, Mazurkiewicz-Bełdzińska M, *et al*. Risdiplam-treated infants with type 1 spinal muscular atrophy versus historical controls. *N Engl J Med* 2021;385:427–35.
8 Iyer AA, Barzilay JR, Tabor HK. Patient and family social media use surrounding a novel treatment for a rare genetic disease: a qualitative interview study. *Genet Med* 2020;22:1830–7.
9 Pacione M, Siskind CE, Day JW, *et al*. Perspectives on Spinraza (Nusinersen) treatment study: views of individuals and parents of children diagnosed with spinal muscular atrophy. *J Neuromuscul Dis* 2019;6:119–31.
10 Kiefer P, Kirschner J, Pechmann A, *et al*. Experiences of caregivers of children with spinal muscular atrophy participating in the expanded access program for nusinersen: a longitudinal qualitative study. *Orphanet J Rare Dis* 2020;15:194.
11 Pasquini TLS, Goff SL, Whitehill JM. Navigating the U.S. health insurance landscape for children with rare diseases: a qualitative study of parents' experiences. *Orphanet J Rare Dis* 2021;16:313.
12 Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. *Int J Qual Health Care* 2007;19:349–57.
13 Burnard P. Teaching the analysis of textual data: an experiential approach. *Nurse Educ Today* 1996;16:278–81.
14 Biogen. New data at cure SMA 2021 highlight the long-term efficacy of SPINRAZA® (Nusinersen) and Biogen's commitment to innovation in SMA therapy. 2021. Available: https://investors.biogen.com/news-releases/news-release-details/new-data-cure-sma-2021-highlight-long-term-efficacy-spinrazar
15 SMA NBS Alliance. Status of spinal muscular atrophy newborn screening in Europe. 2022. Available: https://www.sma-screening-alliance.org/map/
16 Novartis. Newborn screening for spinal muscular atrophy (SMA). 2022. Available: https://www.novartis.com/about/innovative-medicines/novartis-pharmaceuticals/novartis-gene-therapies/newborn-screening-spinal-muscular-atrophy-sma
17 Dangouloff T, Vrščaj E, Servais L, *et al*. Newborn screening programs for spinal muscular atrophy worldwide: where we stand and where to go. *Neuromuscul Disord* 2021;31:574–82.
18 Fortunato F, Rossi R, Falzarano MS, *et al*. Innovative therapeutic approaches for Duchenne muscular dystrophy. *J Clin Med* 2021;10:820.
19 Dowling JJ, D Gonorazky H, Cohn RD, *et al*. Treating pediatric neuromuscular disorders: the future is now. *Am J Med Genet A* 2018;176:804–41.
20 Daniels N. *Just health: meeting health needs fairly / Norman Daniels*. Cambridge: Cambridge University Press, 2007.
21 McMillan HJ, Campbell C. We need a "made in Canada" orphan drug framework. *CMAJ* 2017;189:E1274–5.
22 Rawson NS, Adams J. Access to new drugs for rare disorders in Canada. *CMAJ* 2018;190:E840.
23 Panju AH, Bell CM. Policy alternatives for treatments for rare diseases. *CMAJ* 2010;182:E787–92.
24 Baumbusch J, Mayer S, Sloan-Yip I. Alone in a crowd? Parents of children with rare diseases' experiences of navigating the healthcare system. *J Genet Couns* 2018;28:80–90.