Pieper Ellen (Orcid ID: 0000-0003-4594-8279)

Communication about spinal muscular atrophy and genetic risk within families: An Australian pilot study.

Original article

Full names of the authors and the addresses of the institutions at which the work was carried out:

Ellen Pieper[1]
[1]Department of Paediatrics,
The University of Melbourne,
Parkville VIC 3051

Adrienne Sexton[1,2]
[1]Department of Paediatrics,
The University of Melbourne,
Parkville VIC 3051
[2]Genomic Medicine & Parkville Familial Cancer Centre
The Royal Melbourne Hospital
Grattan St, Melbourne 3050

Monique M. Ryan[1,2,3]
[1]Murdoch Childrens Research Institute
[2]Department of Neurology
The Royal Children's Hospital
50 Flemington Rd, Parkville 3052
[3]The University of Melbourne,
Parkville VIC 3051

Robin Forbes[1,2,3]
[1]Victorian Clinical Genetics Services
[2]Murdoch Childrens Research Institute
[3]Department of Neurology
Royal Children's Hospital
50 Flemington Rd, Parkville 3052

Manuscript, proofs and requests for offprints should be sent to:
Ellen Pieper
Genomic Medicine & Parkville Familial Cancer Centre
The Royal Melbourne Hospital
305 Grattan St, Melbourne 3000
ellen.pieper@mh.org.au
0411 222 926

Email address of the corresponding author: ellen.pieper@mh.org.au

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Conflicts of interest

The authors report no conflicts of interest.

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Abstract

Aim: In families with a child diagnosed with spinal muscular atrophy (SMA), siblings who do not have SMA could still be genetic carriers of the condition. This study is the first to explore how siblings of patients with SMA learn about the condition and their genetic risk.

Method: In-depth, semi-structured interviews were conducted with several parents and unaffected siblings of people with SMA types II and III in Australia. Thematic analysis was performed.

Results: Siblings described learning about SMA gradually over time through conversations with their parents and other sources, including the Internet, biology classes and support groups. Parents and unaffected siblings described challenges in family communication due to the emotional intensity associated with having SMA in the family. Most siblings did not report learning from their family how the inheritance of SMA related to their own genetic carrier risk and possible reproductive implications.

Conclusion: Siblings described their parents as being open and honest in communicating about SMA; however, this study found that communication before the age of understanding abstract concepts, in combination with the emotional intensity of SMA, resulted in gaps in knowledge about SMA.

Key words: adolescent; genetics; communication; genetic counselling; spinal muscular atrophy.

What is already known on this topic?

1. In families where a child has SMA, the unaffected siblings have a 2 in 3 risk of being a genetic carrier of SMA.
2. Family communication to siblings about genetic information can be a difficult process, in which it has been shown that open, honest and gradual communication is most effective.
3. Very few previous studies have explored the experiences of siblings in SMA families.

What this paper adds

1. This study makes the first contribution towards understanding the experiences and support needs of the siblings of patients with SMA.
2. Highlighted is the importance for parents to explain the autosomal recessive nature of SMA to their children at an age when they can comprehend abstract information.
3. Both siblings and parents found it challenging to discuss emotionally difficult or complex topics such as life expectancy, autosomal recessive inheritance and the possible reproductive implications of being a genetic carrier.

Introduction

Spinal muscular atrophy (SMA), the second most common fatal autosomal recessive disease after cystic fibrosis, leads to progressive weakness and muscle atrophy.[1] SMA has an incidence of approximately 1 in 10,000 live births and a carrier frequency of 1 in 41 in Australia.[2] The condition has been traditionally classified according to a spectrum of clinical severity and age of onset: before six months (type I), seven to 18 months (type II), or after 18 months (type III) of age.[3] The International Standards of Care for SMA (2018)[4, 5] updated from 2007[6] established a baseline for best-practice clinical care and outcomes, recommending that patients require care and management from a multidisciplinary team.

In families with a child with SMA, unaffected siblings have a 2 in 3 risk of being a genetic carrier of SMA. Effective family communication allows unaffected siblings to be aware of their genetic carrier risk and learn about their future reproductive options. However, studies in other conditions have shown that family communication about genetics can be a difficult process.[7, 8] Gradual, open and honest communication styles are generally recognised as being more effective and beneficial for a sibling's psychological wellbeing, in comparison to controlled disclosure and non-disclosure.[9, 10]

Notably, gradual communication of risk information allows adolescents to adjust to and cope with their genetic risk whilst maintaining healthy relationships within their family.[11, 12] Furthermore, when genetic information is not communicated, unaffected siblings may form misconceptions that they might develop the disease themselves, feel stressed, experience self-blame or practice risky behaviour.[11]

Forrest et al. (2012) found that only 1 in 6 siblings of patients with SMA undertook carrier testing. They suggested that family communication could be an influential factor in determining whether family members are informed about their genetic risks and undergo genetic testing.[13] Genetic conditions with more complex inheritance patterns, coupled with the high daily intensity of a condition, can negatively impact on the discussion of genetic risk within a family.[14] As patients with SMA require a high nature of care, we expect this may also be the case for SMA.

We aim to explore whether and how siblings learn about SMA, particularly about the genetic implications for themselves. Here, we make the first contribution towards understanding the experiences and support needs of siblings in SMA families.

Methods

A qualitative framework of phenomenology was used, which aims to describe and reflect on an individual's lived experiences. The study team consisted of a student researcher who conducted all interviews, two genetic counsellors, and a paediatric neurologist. This study acknowledges the researchers may have a degree of influence on the research findings, termed reflexivity [15], due to their interest in this topic. The researchers maintained awareness of their perspective as health professionals at all stages of the research process in order to avoid any bias.

There were two sample populations in this study: (i) siblings and (ii) parents of people with SMA types II or III. Purposive sampling was used to recruit unaffected siblings who were aged 15-25, and to recruit parents who had both an affected and unaffected child. Individuals meeting these criteria were identified from patient records. This study did not include families with patients diagnosed with SMA type I, as these siblings may not have known their sibling with SMA type I.

Parents in eligible families were posted a letter of invitation, a Plain Language Statement and a consent form from the patient's treating clinician. Parents were asked to pass the information packs to their eligible children. Participants were offered a choice of either a telephone or face-to-face interview. A flexible question guide with open-ended questions invited participants to discuss specific situations relevant to their experiences of SMA. Parent participants were encouraged to discuss their thoughts and feelings about communicating about SMA to their unaffected children. All interviews were conducted before the availability of Nusinersen (Spinraza) treatment for SMA in Australia.

The interviews were audio-recorded and transcribed verbatim, with pseudonyms used. The transcripts were not returned to the participants for checking. Interviews were analysed using thematic analysis[16]. Initially, one researcher familiarised themselves with the transcripts in detail and identified broad themes and subthemes through an iterative coding process. Three researchers independently coded the transcripts and later compared and discussed the proposed themes to triangulate the analysis. The transcripts were coded with the assistance of QSR NVivo 11.4 software.

Results

A total of 11 families were eligible to participate in the study. Of these, two families declined participation, and three families did not respond to the letter of invitation. The participants comprised (i) four siblings from three families and (ii) three parents from three families, resulting in a total of seven interviews across six families, presented in Table 1 and Table 2. There were five families with SMA type II and one family with SMA type III. Sibling participants were aged 16 to 25 years, and parent participants had unaffected children between the ages 3 to 7 years. Six interviews were conducted by telephone, and one interview was conducted face-to-face. The duration of the interviews was between 27 – 55 minutes, with an average of 44 minutes. Due to the low sample size, data saturation was not sought. Themes relating to sibling experiences and the times of communication about SMA are presented.

Theme 1: Learning about SMA in "bits and pieces."

Siblings described their first realisation that their affected brother or sister had SMA (Subtheme 1: Table 3). This experience occurred between the ages of 6 and 7, where most participants (3/4)

questioned why their affected sibling was different or in a wheelchair. This prompted siblings to ask their parents about SMA. All sibling participants in the study (4/4) reported having an early, brief conversation about the inheritance of SMA between ages 8 to 13(Subtheme 2: Table 3, Fig. 1). The focus of these conversations was to explain the reason their affected sibling had inherited the condition, rather than to explain the genetic risk for the unaffected sibling. One participant felt that the genetic cause of SMA did not feel relevant at the time it was explained. Sibling participants did not recall ongoing conversations with their families about their genetic risk.

Siblings described learning about SMA gradually from many sources, including their parents, health professionals, the Internet, biology classes and textbooks, booklets, television programs and SMA support organisations (Subtheme 3: Table 3). Often, family communication was prompted by a significant life event; for example, the sibling with SMA undergoing surgery or following the initiation of a new relationship (Subtheme 4: Table 3). One participant described that his parents provided him with age-appropriate information over time. Two participants attended genetic counselling appointments at ages 23 and 25 (Subtheme 5: Table 3). Both described their genetic counselling sessions as being informative, whereby the reproductive options and implications of being a genetic carrier were discussed.

Theme 2: SMA is "hard to talk about."

While siblings generally described that family communication about SMA was welcomed, all sibling participants explained that SMA was sometimes challenging to discuss due to the emotional intensity of these conversations. All described a significant period in their lives when they realised the severity or prognosis of SMA, between 16 to 22 years of age (Subtheme 1: Table 4). Those who were aware of the prognosis (3/4) described a sudden period of anticipatory grief, and described feeling devastated and helpless (Subtheme 2: Table 4). Participants used the phrases "took a toll", "couldn't stop thinking about it", and "hit home" in describing their period of anticipatory grief. All said that they ultimately felt their relationship with their brother or sister with SMA had strengthened over this period.

The severity of SMA made it challenging for some siblings to initiate conversations about SMA, with the concern that they could make their parents feel uncomfortable (Subtheme 3: Table 4). For two

participants, this led to independent information seeking using the Internet. Notably, one sibling described that she had formed misconceptions about the SMA subtypes.

Theme 3: Parents intended to explain SMA carrier risks between ages 8-14

All parents in the study (3/3) intended to communicate with their unaffected children about SMA, including their genetic carrier risk. Parents explained that their children had not yet reached an understanding of SMA between 3-7 years of age. Parents had clear intentions of how they planned to communicate about the genetics of SMA to their unaffected children (Subtheme 1: Table 5, Fig. 1). All parents intended to initiate a discussion about the cause of SMA from 6 years and to explain the risk of being a genetic carrier from 8 to 14 years of age. Parents planned to impart information about SMA in an open, gradual manner and considered both the age and maturity of their children when planning how to discuss SMA (Subtheme 2: Table 5). Parents chose appropriate language that their children would understand; for example, one parent consciously did not use the term "disease" as she did not think her child currently understood the word. Communication about SMA was difficult at times to balance the parental roles of both protecting and informing their children (Subtheme 3: Table 5). One parent anticipated difficulty in initiating a discussion about life expectancy in SMA because she did not want her children to feel scared.

Theme 4: Public awareness of SMA as the first step towards better support.

All participants were encouraged to consider what additional support could be helpful for siblings in SMA families. Most participants (5/7) had actively engaged with support organisations, including sibling support days. Most participants, including both siblings and parents (5 of 7), expressed that they would like greater public awareness about SMA (Subtheme 1: Table 6). In these cases, participants hoped for greater understanding and sensitivity from the public, which would also make it easier for families to reach out for social support. Other suggestions for support included: more psychosocial support for the person with SMA, age-appropriate communication resources and additional help from health professionals (Subthemes 2, 3 and 4: Table 6)

Discussion

The communication of genetic information in families is frequently described as a process, rather than an event in time.[17] A study by Metcalfe et al. (2011) explored the ages that children come to an understanding of genetic information. The present study identified similar stages of learning about a genetic condition in siblings: noticing visual differences, learning about inheritance, learning about prognosis, and learning of the reproduction implications for themselves. It is common for the first conversations about inheritance to be about shared physical traits within the family, such as hair colour or freckles[18], as was found in the present study. In this study, parents communicated the concept of autosomal recessive inheritance from age 6 by explaining that both mother and father carry a faulty gene, which is passed on to the child with SMA. This definition describes how the affected child came to have SMA but does not suggest there may be genetic implications for other family members. Parents in this study intended to communicate about the reproductive implications of SMA from 8-14 years of age, which is younger than the age at which most children can understand these implications (15-17 years).[11] These results are similar to a study about families affected by cystic fibrosis, where parents also intended to disclose their children's carrier risk at an early age (6-11 years).[19] One sibling in the present study highlighted that the concept of inheritance did not feel relevant at the time it was explained to them. At these younger ages, children may not have connected the concept of inheritance with an impact on their reproductive health.

All participants described having an open relationship with their parents. Parents also encouraged family communication and were available to answer their children's questions. An open style of communication can empower an individual to feel more in control of his or her health and provides autonomy so that children can make informed health and reproductive decisions.[8] Siblings also described that communication was elicited by life events, which has been previously described for other genetic conditions.[20] Parents were specific about when they intended to initiate conversations; however, participants in the study did not describe remembering many conversations that their parents had independently initiated. One possibility is that the experience of initiating conversations about SMA is more complicated than anticipated. It is also possible that the prompted nature of some conversations was more memorable to siblings during interviews. Two participants experienced a period where they used the Internet to learn more about SMA. While the Internet can be an easily accessible source of information, it can also be a complicated and overwhelming source for young people and a potentially inaccurate source of health information.[21]

Gradual communication is a commonly described style of disclosure in families affected by chronic illness.[8] This style of communication allows adolescents to adjust and discuss each new piece of information with their parents. Siblings in the present study described that the information they learnt gradually was predominantly about the clinical care needs of their sibling with SMA. It was evident that some topics about SMA were more difficult to discuss within the family due to their emotional intensity. Previous studies have described that parents face a dilemma to protect their children from feeling afraid about a genetic condition while also informing them of the condition[22]; a dilemma expressed by one parent in this present study. Passive nondisclosure is a concept whereby parents do not directly withhold information from their children, but due to the family dynamic, less disclosure occurs.[23]

A major subtheme that arose was the effect of learning about the prognosis of SMA. The majority of siblings came to a realisation about the prognosis between ages 16-22. Parents did not intend to disclose the life expectancy of SMA to their children, however it is clear that siblings become aware of this over time. One parent explained that this was due to the emotional difficulty of the conversation and that she didn't want her children to feel afraid of their sibling's mortality. Siblings found difficulty to come to terms with the prognosis of SMA and experienced a period of anticipatory grief. Similar themes have also been identified in a recent study exploring communication to the sisters of patients with DMD.[20]

Despite SMA being one of the most common fatal genetic conditions, most participants described there is little public awareness about SMA. They suggested that greater awareness would help the family as a whole. The lack of public awareness of SMA may put family members at risk of experiencing disenfranchised grief- grief that is not socially recognised.[24] Parents in the current study also hoped for more age-appropriate resources to communicate about SMA in a more positive light. Together, these findings suggest that more public education and resources could be helpful for SMA families. It may be beneficial for parents to have a conversation with a health professional about family communication where the main focus is not solely on the child with SMA.

Due to the small sample size of the study, data saturation was not sought, therefore the results are not generalisable and this is a limitation of the study. Member checking of the transcripts did not occur, and while every effort was made to accurately represent participant views, this is another potential

limitation of the study. Here, we provide the first insight into the way siblings learn about SMA and their genetic risk in Australian families. Therefore, the findings are not intended to be generalisable to all families with SMA but raise themes that may be relevant to many families. We have found that, in some families, there may be difficulties communicating about SMA due to the emotional intensity of SMA. Thus, this is an area where additional research on a larger scale would be beneficial.

Overall, the findings of this study are in line with the notion that the disclosure of information about genetic disease is a multi-stage process, where details are shared over time and siblings experience multiple stages of coping and understanding.[10, 22]

Conclusion

This study provides unique and valuable insight into the experiences of siblings of patients with SMA types II and III. Siblings in this study described gradual, open and honest communication in their family; however, both siblings and parents faced challenges in communicating about emotionally difficult subjects such as life expectancy, autosomal recessive inheritance and the reproductive implications of SMA. This study also highlights the importance of communicating complex information, such as genetic carrier risk, at an age that children can comprehend the implications for their future. With the introduction of new treatments for SMA prompting the widespread introduction of newborn and population screening programs, carrier-testing discussions may become more prevalent. With this, there must be further research into how to support siblings with a high carrier risk for SMA.

Conflict of interest: The authors report no conflicts of interest.

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(Figure_1.pdf)

Figure 1. a) The ages that children understand genetic information, as found in Metcalfe et al. (2011); b) The ages that unaffected siblings in the present study learnt about SMA; c) The ages that parents in the present study intended to disclose information about SMA to their unaffected children. *'Visual differences'* refers to the physical presentation of a genetic condition; *'inheritance'* refers to the inheritance of the genetic condition, *'prognosis'* refers to the prognosis and life expectancy of a person with a genetic condition, *'familial implications'* refers to the possibility and meaning of being a gene carrier and the reproductive options available.

Table 1 Demographic characteristics of the sample: sibling participants

Sibling	Family (A-C)	Age (years)	Sex	SMA type (proband)	Interview method
1	A	16	Female	Type II	Face-to-face
2	B	18	Male	Type II	Phone
3	C	23	Male	Type II	Phone
4	C	25	Male	Type II	Phone

Table 2 Demographic characteristics of the sample: parent participants

Parent	Family (D-F)	Age of unaffected child (years)	Mother or father	SMA type (proband)	Interview method
1	D	3, 7	Mother	Type II	Phone
2	E	3	Father	Type III	Phone
3	F	7	Mother	Type II	Phone

Table 3 Theme 1: Learning about SMA in "bits and pieces"

Subtheme	Representative quotation
1. Noticing their siblings' presentation of SMA	*In Primary school growing up … that's when it really hit me when other kids … refer to him as 'oh, your brother, the boy in the wheelchair'. Just stuff like that was when I thought, like wow he really is different.* Sibling 1, 16 yrs.
2. Learning a brief genetic explanation of SMA	*I said, you know, like, 'what actually causes SMA?' And [Mum and Dad] were like, 'oh, well Mum has a bad gene, Dad has a bad gene and they come together and um create SMA.' … I think it would have been just like a brief moment where I was maybe wondering and then they told me it.* Sibling 2, 18 yrs.
3. Learning gradual and age-appropriate information over time	*They always told us everything but I'd say just a bit dumbed down if we were younger and to feed a bit more information to us as we got a bit older.* Sibling 3, 23 yrs.
4. Learning is prompted by life events	*My brother was scheduled to have surgery then and Mum kind of sat me down and told me what was happening.* Sibling 1, 16 yrs.
5. Learning through genetic counselling	*The genetic advisor lady was explaining how it works. I think I might be able to remember there's one carrier in 1 in 35 chance and if you're both carriers there's a 1 in 4 chance or something.* Sibling 4, 25 yrs.

Table 4 Theme 2: SMA is "hard to talk about"

Subtheme	Representative quotation
1. Coming to a sudden realisation about the prognosis of SMA	*I might've been told early on but I didn't realise how apparently when my sister was born, she was told she might not live until she was 19 or something…so that was one thing that sort of hit home with me about a year ago when I found out, well when I sort of realised and really thought about it.* Sibling 3, 23 yrs.
2. Experiencing anticipatory grief for sibling with SMA	*I really couldn't stop thinking about it just to know he is such a big part of my life and that yeah he might not be here that much longer. I just found it really hard just even to say [sibling's name] without you know, like triggering an emotional response.* Sibling 2, 18 yrs.
3. Difficulty communicating about SMA due to emotional discomfort	*I thought um, I just wanted to like – with Mum I didn't want to make her upset like, it wouldn't make her upset but I didn't want to put her in an uncomfortable position.* Sibling 1, 16 yrs. *We'll try and not go into too much detail and stuff just because [SMA] is a bit hard to talk about… Definitely gets pretty hard to talk about, probably just how severe it is, I guess.* Sibling 2, 18 yrs.

Table 5 Theme 3: Parents intend to communicate about SMA, including genetic carrier risks

Subtheme	Representative quotation
1. Providing a brief genetic explanation	*I think I did try to explain briefly to [unaffected child] that Mummy and Daddy have a faulty gene and unfortunately sometimes it will pass onto their babies when the mum carries. I think he's still too little to understand the genetic side.* Parent 1
2. Providing information gradually over time	*I will give him information time to time as he will be getting older… it's day by day.* Parent 2
3. Difficulty discussing the prognosis of SMA	*We haven't really harped on very much about um the risk of him dying early or any of that, and partly because I don't want to scare [affected child] and have him wondering every day when it's going to happen. But, I guess it's the same with [unaffected child], I don't want to scare him either.* Parent 3

Table 6 Support needs for siblings, identified by participants

Subtheme	Representative quotation
1. Increased public awareness about SMA	*There's not enough information out there or well, like being told much to the public, so they wouldn't really understand what SMA is."* Sibling 3, 23 yrs.
2. Practical and emotional support for the sibling with SMA	*I've noticed that my sister would go through phases where she gets really upset. . . Like all the feelings would come up that she wasn't quite the same as everyone else . . . it would affect Mum and Dad 'cos they'd be seeing her upset and then they'd get upset, sort of keep that negative energy going around . . . and obviously got a bit of a flow on effect on the rest of us if we're feeling sorry for her, and you're sort of helpless, you can't do anything about it . . . It could just be something as simple as – she wanted to get into a café and she couldn't get in because there's no ramp . . . Just little things like that can make her life a bit easier, to keep her mental state healthy; it would just make life a bit easier. It doesn't take much.* Sibling 3, 23 yrs.
3. Learning resources about SMA for children	*I think at this point videos are a really good way [for children to learn about SMA]. There are some good descriptive videos online about how the disease itself manifests and how the treatment works and all the rest.* Parent 3
4. Support from health professionals	*To have an option of coming in as a whole family and to have someone help us explain it would be a great thing. Because there will always be questions that they'll answer that we can't answer.* Parent 3

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Original article

Full names of the authors and the addresses of the institutions at which the work was carried out:

Ellen Pieper[1]
[1]Department of Paediatrics,
The University of Melbourne,
Parkville VIC 3051

Adrienne Sexton[1,2]
[1]Department of Paediatrics,
The University of Melbourne,
Parkville VIC 3051
[2]Genomic Medicine & Parkville Familial Cancer Centre
The Royal Melbourne Hospital
Grattan St, Melbourne 3050

Monique M. Ryan[1,2,3]
[1]Murdoch Childrens Research Institute
[2]Department of Neurology
The Royal Children's Hospital
50 Flemington Rd, Parkville 3052
[3]The University of Melbourne,
Parkville VIC 3051

Robin Forbes[1,2,3]
[1]Victorian Clinical Genetics Services
[2]Murdoch Childrens Research Institute
[3]Department of Neurology
Royal Children's Hospital
50 Flemington Rd, Parkville 3052

Manuscript, proofs and requests for offprints should be sent to:
Ellen Pieper
Genomic Medicine & Parkville Familial Cancer Centre
The Royal Melbourne Hospital
305 Grattan St, Melbourne 3000
ellen.pieper@mh.org.au
0411 222 926

Email address of the corresponding author: ellen.pieper@mh.org.au

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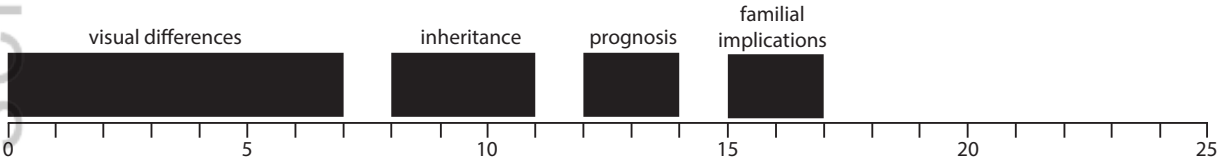
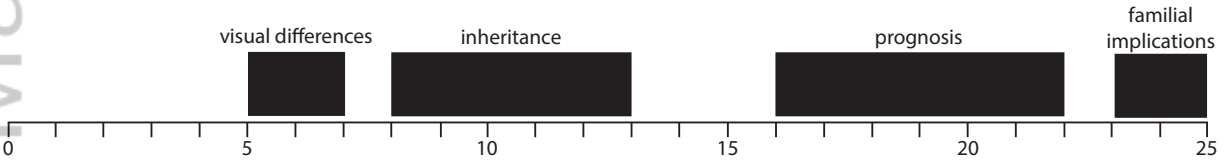
a) The ages that children understand genetic information



b) The ages that siblings in this study learnt about SMA



c) The ages that parents in this study intended to disclose information about SMA