Living with **Spinal Muscular Atrophy** in Malaysia

A STUDY

#thinkSMALLactBIG

We Care Journey

WeCareJourney is dedicated
to the treatment of
spinal muscular atrophy (SMA),
the number one generic
cause of death of infants.
An SMA diagonosis can be overwhelming,
and we give patients and families
the information and resources they need to
live active, engaged, and hopeful lives today.
We are commited to respecting
and fostering diversity and
inclusion in the people we help.

***A special thank you to Ainaa Farhanah Amali**, a graphic designer living with SMA ,
for her cover and graphic contribution in the designing of this book.*

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Project Team

Research Team

Dr Ch'ng Gaik Siew
Consultant Clinical Geneticist,
Department of Genetics
Penang Hospital

Dr Azlina Bt Ahd Annuar
Senior Lecturer & Neurogeneticist,
Department Of Biomedical Science
Faculty of Medicine, University Malaya

Dr Fahisham Taib
Senior Lecturer & Paediatrician
Hospital Universiti Sains Malaysia

Dr Karina Koh
Medical Officer/Research Coordinator
Clinical Research Centre (CRC),
Kuala Lumpur Hospital

Celine Koh Cha Ling
Registered Counsellor

Lee Boon Hock
Registered Counsellor

Advisor

Dr Ling How Kee
Social Work Educator and
Research Consultant

Coordinator

Edmund Lim Soon Chin
President of Persatuan
WeCareJourney and Spinal
Muscular Atrophy and Disability
Activist and Advocate

Acknowledgements

I remember hoping that what the doctor was going to tell us about our child's diagnosis wouldn't change the lives of our family. The doctor started describing Spinal Muscular Atrophy (SMA), but I really wasn't following the explanation - something about being severely disabled and the condition getting progressively worse; that there was no cure nor treatment (then) and we should prepare for the worst. As a parent, I am sometimes asked what's the biggest obstacle to overcome in raising a child with a progressive life-limiting rare condition like SMA. We have a terribly long list of burdens, and we need help to overcome many obstacles. Some people follow up to ask "how can I help?". And so, over time, as I got to know more families with SMA, the importance of giving everyone the chance to tell their stories, thoughts and feelings, and the realities of the many challenges they face, became apparent to me.

This report was put together by a team of passionate and hardworking doctors and counsellors, who were gathered and led by a very compassionate and dedicated clinical geneticist, Dr. Ch'ng Gaik Siew. The contributions of social workers Luei Jia Qi in the preparation for this project and Suzanah Abd Hamid in helping to conduct some of the earlier interviews was also invaluable. The research team has worked tirelessly to raise the profile of SMA in Malaysia by documenting the experiences of patients and caregivers in the hopes of bringing positive change to their lives. They were all touched by the patients' and caregivers' stories of perseverance, tenacity, strength and unyielding spirit in the face of disappointments and hopelessness.

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Some people told us this project could not be done. Others said it was naive to try and change the situation. But we should always believe that we can make a change. We hope that all families and stakeholders will take from this report the knowledge and insights needed to help create better living experiences for families with SMA in Malaysia.

Thank you!

Mr Edmund Lim
President of Persatuan WeCareJourney
And Spinal Muscular Atrophy and Disability
Activist and Advocate

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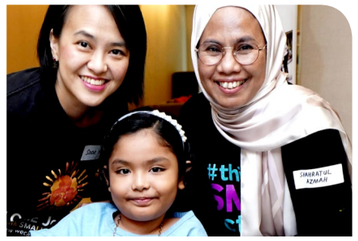


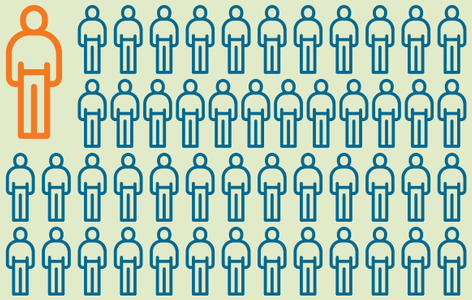
List of Abbreviations

BiPAP	Bilevel Positive Airway Pressure
CPAP	Continuous Positive Airway Pressure
DASS	Depression, Anxiety and Stress Scale
FGD	Focus Group Discussion
ICU	Intensive Care Unit
IDI	In-Depth Interview
IEP	Inclusive Education Programme
IJN	Institut Jantung Negara (National Heart Institute)
JPA	Jabatan Perkhidmatan Awam (Public Service Department)
MOH	Ministry of Health
MREC	Medical Research Ethics Committee
NBS	Newborn Screening
NGO	Non-governmental organisation
OKU	Orang Kurang Upaya (Persons with disability)
PIBG	Persatuan Ibu Bapa dan Guru (Parent-Teacher Association)
PGD	Pre-implantation genetic diagnosis
PND	Prenatal Diagnosis
PWCJ	Persatuan WeCareJourney
PWSMA	Persons with Spinal Muscular Atrophy
QOL	Quality of Life
SMA	Spinal Muscular Atrophy
SMN	Survival Motor Neuron
UNICEF	United Nations Children's Fund

Executive summary

Spinal muscular atrophy (SMA) is a recessively inherited neuromuscular disorder that affects the motor nerves, resulting in progressive muscle weakness, which impacts physical activities such as breathing, swallowing, crawling/walking, head control and overall movement. This can lead to decreased life expectancy in the most severe type, as well as a gradual decline in the patient's ability to perform activities of daily living like eating and bathing. Patients also face an increased risk of pneumonia and difficulties in breathing during sleep. SMA is classified into four clinical types based on the age of onset and the maximum motor function achievable. Brain and sensory functions are not affected.

On average, one in every 50 persons carries mutations in the survival motor neuron (SMN) gene that causes SMA and can pass it on to their children. SMA occurs when a child receives mutated copies of the gene from both parents. It is estimated that one in every 10,000 live births worldwide is diagnosed with SMA. Based on the national birth rate of 500,000 babies a year, this means that there are an estimated 50 babies with SMA born annually in Malaysia.



Estimated
1 in **50** persons
carries the mutated
gene that causes SMA

Currently, there is no cure for SMA, but there are therapies that can delay clinical progression and relieve certain symptoms. The differences in presentation of the condition often results in delays and difficulties in diagnosis, and contributes to the challenges in understanding the distribution of the disease. The impact of this chronic debilitating condition can be manifold, affecting patients and their family members in various ways, including psychologically, financially due to the high cost of medical care and possible loss of income, and socially due to lack of educational and employment opportunities.

50 babies with SMA are born annually in Malaysia

In Malaysia to date, no studies have been done to document the experiences of SMA patients, their families and caregivers. The objective of this study is to fill this gap and to gain an understanding of the impact of living with SMA from the perspectives of persons with spinal muscular atrophy (PWSMA) and their caregivers. This cross-sectional study consisted of two parts: a quantitative study where validated questionnaires were distributed to all identified participants, and a qualitative study where in-depth interviews (IDIs) and focus group discussions (FGDs) were conducted based on an interview guide. Eligible participants were recruited using a purposive sampling method. A total of 42 individuals participated in the quantitative survey, 13 of whom were PWSMA and 29 who were caregivers. After completing the quantitative survey, seven PWSMA and 23 caregivers continued on with the IDIs and FGDs.

"The impact of this chronic debilitating condition can be manifold, affecting the patients and their family members in various ways, including psychologically, financially and socially."

Of the 13 PWSMA, five were male and eight were female. Their age ranged from 11 to 42 years. Nine had SMA type 2 and four had SMA type 3. The majority had completed tertiary education. Six were in full- or part-time employment, drawing an income of between RM800 to RM2,000.

Eleven male and 18 female caregivers, who were mostly family members of the PWSMA, participated in the study. Eight were caregivers to PWSMA type 1, 11 were caregivers to PWSMA type 2, and 10 were caregivers to PWSMA type 3. The majority had also completed tertiary education.

A large portion of PWSMA and caregivers reported experiencing stress, anxiety and depression while coping with SMA. The PWSMA also reported that the symptoms that most significantly affected their daily lives were muscle weakness and joint contractures, followed by lung infection and fatigue. This was mirrored by the observations of the caregivers. While visits to various specialist clinics are common, none of the participants had ever met with a mental health professional despite the presence of mental health issues. Of concern was the finding that the majority of the participants were unaware of clinical trials related to treatment for SMA, as well as preimplantation genetic diagnosis (PGD) and newborn screening (NBS) tests for SMA.

“A large portion of PWSMA and caregivers reported experiencing stress, anxiety and depression while coping with SMA.”

The IDIs and FDGs uncovered various themes pertinent to the impact of living with SMA. Among them were experiences related to diagnosis, including that of dismissive healthcare professionals; lack of information and supportive services; and a long and tortuous journey to getting the diagnosis for many. It was also clear that self-doubt and inner turmoil were common experiences, with many having to cope with changing lifestyles and family relationships, diminished social lives, and financial burdens. Another theme that surfaced was the experience of an uncaring and unsupportive schooling environment with some incidences of imposed isolation and bullying.

When discussing the participants’ future hopes and wishes, themes such as the desire for access to treatment and clinical trials; the need for holistic care post-diagnosis, including provision of palliative care; professional mental health counselling; a respite care system; and support groups, were raised. A call to the government to increase awareness of and improve medical care services for SMA, as well as improve access for disabled persons, particularly those with wheelchairs, in public places and schools, was also recorded.

Recommendations:

This study has highlighted a plethora of issues and challenges experienced by PWSMA and their caregivers, providing evidence to inform service provision and policy formulation to better meet their needs and rights. Recommendations based on the findings encompass several aspects, such as strengthening clinical management for rare diseases, including implementation of a systematic approach to diagnosing SMA; involving a multidisciplinary team (MDT) for better standards of care; looking into specialised pulmonary, orthopaedic and rehabilitative care; and providing opportunities to participate in clinical trials and receive novel therapies. Additionally, recommendations to improve psychosocial support services, for example, empowering patient-parent advocacy organisations, reforming educational institutions and ensuring equality in employment opportunities, were made.

Conclusion:

A call to policymakers and insurance providers to ensure a clear and committed policy direction, covering regulations and incentives, and the provision of genetic non-discrimination insurance, is also being made. Lastly, increasing community education and awareness, as well as future research on SMA, is also recommended.

Overall, the sustained efforts from all relevant stakeholder such as the government, policy makers, healthcare providers and workers, medical technology and pharmaceutical industries, assistive technology industry, insurance industry and other funders, Non-governmental organizations, PWSMA and their caregivers are required to bring about systemic change and lessen the burden of living with SMA in Malaysia.

Chapter 1
Introduction

1.1

Background

Spinal muscular atrophy (SMA) is a recessively inherited neuromuscular disorder caused by deletions or mutations in the survival motor neuron 1 gene (SMN1) (Lefebvre 1991). On average, one in every 50 people is a carrier of mutations in this gene. A prevalence of approximately one to two per 100,000 persons and an incidence of around one in 10,000 live births have been estimated with SMA type 1 (the most severe form) accounting for around 60% of all cases (Ingrid 2017). Based on the birth rate of approximately 500,000 babies a year, an estimated 50 babies with SMA are born every year in Malaysia.

SMA is diverse in presentation and is classified into clinical types based upon the age of onset (which is related to disease severity and survival) and the maximum motor function achievable. Type 1 is the most severe and life-threatening form. Patients with SMA type 1 are unable to sit without support and typically do not live beyond two years of age without respiratory support. Type 2 patients cannot stand or walk on their own, but are able to sit independently, while type 3 are patients whose symptoms start during childhood and are initially able to walk. Type 4 are patients whose symptoms start during adulthood. The spectrum of severity has been associated with the copy number of the SMN2 gene. The SMN2 gene is almost identical to the SMN1 gene, except that most of the SMN protein it encodes is truncated and rapidly degraded.

> “Currently, there is no cure available for SMA, but there are therapies that can help to delay progression and relieve some symptoms.”

The burden of disease, treatment and support needs of people with different types of the disease are distinct. Currently, there is no cure available for SMA, but there are therapies that can help to delay progression and relieve some symptoms. The diversity of the presenting symptoms can lead to delays and difficulties in diagnosing SMA, especially outside of specialist clinics, and contributes to the challenges in understanding the epidemiology of the disease. In addition, the impact of this chronic debilitating condition can be manifold, affecting the PWSMA and their family members in various ways, such as psychologically, financially due to high cost of medical care and possible loss of income, and socially due to lack of educational and employment opportunities. Stigmatisation by the public towards PWSMA may not only affect the mental health of the PWSMA, but also that of their parents, which may in turn affect their ability to care for their child.

There is also a lack of knowledge among the general public and healthcare professionals about the health challenges and psycho-socio-economic issues faced by PWSMA. Often, these issues and experiences have a deep impact on the PWSMA and their families, and are shared verbally or through mass or social media, but are not properly documented otherwise. Currently, there is no research that accurately documents the experience and impact SMA has on patients and families in Malaysia.

Obtaining patient feedback and their experiences with the healthcare system has been shown to subsequently affect hospital practices and clinical care plans in a positive manner for such patients (Anhang 2014). It can be used to reflect clinical and healthcare quality measures and monitor if the principles of person-centred care are being followed. Person-centred care is one of the 13 fundamental standards of care that the Care Quality Commission (the independent regulator of health and social care in England) requires healthcare providers to uphold. Furthermore, patient surveys are essential to identify potential areas of improvement, including management of patient expectations and performance of general practitioners (Boiko 2016).

Recently, UNICEF published a report detailing the issues experienced by children living with disabilities, which includes those with SMA (UNICEF 2017). While this was an important first step to raising the profile of children with various types of disabilities, little is known specifically about children and adults with SMA, as well as their caregivers. These patients and their caregivers may face issues and challenges unique to their condition. This study aims to describe the impact of living with SMA from the perspective of patients and caregivers in Malaysia.

1.2

Objectives

1.2.1 General Objective

To study the impact of living with SMA in Malaysia from the perspective of patients and caregivers.

1.2.2 Specific Objectives

- To explore the impact of SMA on the daily activities of PWSMA and their caregivers.
- To examine the symptoms that affect PWSMA the most from the point of view of PWSMA and their caregivers.
- To elucidate the thoughts of PWSMA and their caregivers on the management and treatment options for SMA.

Chapter 2
Study Methods

2.1

Study Design

This study utilised a cross-sectional study design. It consisted of two parts: the first part was a quantitative study where questionnaires were distributed to all identified participants (Appendix 1 & 2), and the second part was a qualitative study utilising in-depth interviews (IDIs) and focus group discussion (FGD) sessions based on an interview guide (Appendix 3).

2.2

Participants and Sampling Criteria

Participants of the study were all Malaysian PWSMA and their caregivers. Recruitment was through collaboration with Persatuan WeCareJourney (PWCJ), a support group for PWSMA and caregivers or parents of children and adults with SMA.

The criteria of inclusion were :

- **For patients:** Confirmed diagnosis of any type of SMA through clinical evaluation and/or genetic testing.
- **For caregivers:** Provides a minimum of four hours per day of care or help with at least one activity of daily living to a PWSMA (America Psychological Association).
- Willing and able to provide written and signed informed consent, or in the case of those under 18 years of age, provision of signed informed consent by their legally-authorised representative after the nature of the study has been explained to them.

2.3

Study Tools

For the quantitative component, the MySMA Quantitative Questionnaire was used. This questionnaire was adapted from Cure SMA's Voice of the Patient Report 2018. It consists of questions meant to elicit the respondent's demographic details, activities of daily living, disease history and treatment (Appendix 1). To further gauge the psychological state of the participants, the instrument DASS 21 (Depression, Anxiety and Stress Scale - 21 items) was included as part of the questionnaire (Appendix 2).

The qualitative part of the study involved the participants in an IDI or FGD using a semi-structured interview guide titled MySMA Qualitative. The interview guide is an open-ended set of questions exploring the impact of SMA on the daily living activities of patients and their caregivers, the symptoms that matter most to them, their worries and concerns, as well as their perspectives on the management and treatment options for SMA (Appendix 3).

Both the MySMA Quantitative Questionnaire and MySMA Qualitative were subjected to a translation and validation process. Consent was obtained from the original authors for the questionnaires' adaptation to the Malaysian setting. The questionnaires were then translated into Bahasa Malaysia and Mandarin, and validated prior to the start of research.
Native speakers of Bahasa Malaysia and Mandarin did their respective forward-backward translations from English. Once completed, an expert committee (consisting of two experts on SMA, the original translators and an unbiased bilingual translator not involved in the original translation) produced the final version of the translated questionnaires.
The questionnaire was then subjected to face validation by five people, who were selected randomly from attendees of the Genetic Clinic in Kuala Lumpur Hospital and were not included as actual participants of the research. All changes suggested by the participants were reviewed by the expert committee to produce the final translated versions of the questionnaire. The study proposal, which included the questionnaire and interview guides, was also approved by the Medical Research Ethics Committee (MREC) of the Ministry of Health (MOH), Malaysia.

Before the commencement of the study, a capacity-building workshop was held to familiarise the team researchers and research assistants with the objectives of the study, fine-tune the questionnaires, and clarify the concepts and terms in both Bahasa Malaysia and Mandarin. The team adviser, who has expertise in qualitative research, highlighted the major issues and guiding principles to be observed in qualitative research.

2.4

Informed Consent and Withdrawal Process

Potential participants were contacted via email or the WhatsApp messaging app. The Participant Information Sheet and Informed Consent form were attached in the first email or WhatsApp message for them to read. This was to ensure that they knew all the details about the study before giving consent to participate. The contact details of the researchers were also provided to give them ample time and opportunity to clarify any queries they had before deciding whether or not to participate.

After reading the information, those who agreed to participate proceeded to sign the Informed Consent form (or Assent Form if they were below 18 years of age) and were then contacted to set up an appointment at their convenience to complete the questionnaires and interview.

As participation in this study was entirely voluntary, the participants were able to withdraw from the study at any time. They were allowed to terminate their participation without providing any reason to the researchers. The researchers also did not try to coerce them to complete this study in any way.

2.5

Data Collection

Data collection was conducted between April to December 2019. The questionnaires took about an hour to be completed, while the IDIs lasted one to one-and-a-half hours and the FGDs two to two-and-a-half hours. The IDIs and FGDs were conducted by the team of researchers and as much as possible in the native language of the participants. Each of the interviews was audio-recorded and transcribed verbatim for analysis. In instances where there were gaps in data on the PWSMA, secondary data from case notes and personal communication kept by PWCJ were used to supplement the data with the permission of patient.

2.6

Ethical Considerations and Confidentiality

This study adhered to ethical principles in accordance with the Malaysian Good Clinical Practice Guidelines. Ethical approval from the MOH's MREC was obtained prior to conducting this study (NMRR-18-3248-42367).
As this was a cross-sectional study, there were no inherent risks to the participants that were greater than that of routine activities of daily living. During the course of IDIs, participants who experienced psychological and emotional distress due to the questions asked were given support by the trained counsellors in the research team.

The participants might indeed have benefited directly from having their voices and concerns heard. Better knowledge about SMA, as can be provided for through this study, would be useful for future public awareness campaigns, and to help shape the policies and practices in areas such as treatment options, standards of care, genetic counselling and testing, and education and employment opportunities, thus benefiting all in the SMA community.

Participants' personal details were kept strictly confidential in a private password-protected database. Each participant was given a research identification number used only for this study. There were no personal identifiers attached to the data. All data was entered into a password-protected computer. All paper copies of the data and reports were kept in a secured archive in the investigator's office. The data was kept until the completion of the research and destroyed afterwards. Any publication and presentation of the results of this study will not contain any respondent identifiers and their confidentiality will be protected at all times.

2.7

Data Analysis

Descriptive statistics was used to describe the demographic and characteristics of the participants.

The interview transcripts were read by the researchers, and core concepts and emerging themes were identified. The researchers then jointly generated a set of agreed themes according to the main domains identified in the IDIs. Subsequently, a narrative summary of individual IDIs was constructed to identify the overall themes according to the main domains. These themes were further identified, discussed and interpreted by the researchers. Finally, the salient findings across all interviews were synthesised and the main unifying themes that characterised the participants' experiences and views were identified.

2.8

Methodological limitations

As this study is cross-sectional, findings from this study were mainly descriptive. These findings are also unable to be generalised.

Chapter 3
Results

3.1

Quantitative findings

3.1.1 Demography

A total of 42 individuals responded to the quantitative survey. Thirteen were PWSMA, comprising eight females and five males, who ranged in age from 11 to 42 years, with the majority above 18 years old. Nine had SMA type 2 and four had SMA type 3. The remaining participants were caregivers of PWSMA, comprising 18 females and 11 males. Eight of their children had SMA type 1, 11 had SMA type 2 and 10 had SMA type 3. One non-Malaysian caregiver has been a personal assistant to a type 2 PWSMA for 10 years (Table 1).

Table 1: Distribution of Participants by Sociodemographic Background (n=42)

Characteristics	PWSMA N = 13 (%)	Caregivers of PWSMA N = 29 (%)
Gender		
Male	5 (38)	11 (38)
Female	8 (62)	18 (62)
Ethnicity		
Malay	7 (54)	22 (76)
Chinese	5 (38)	5(17)
Indian	1 (8)	1 (3)
Others	0	1 (3)
Age (years)		
10-19	2 (14)*	-
20-29	9 (69)	2 (7)
30-39	1 (8)	10 (34)
40-49	1 (8)	9 (31)
50 and above	0	8 (28)
SMA type		
Type 1	0 (0)	8 (28)
Type 2	9 (69)	11 (38)
Type 3	4 (31)	10 (34)
Highest formal education		
No formal education	1 (8)	1 (3)
Primary	4 (31)	0 (0)
Secondary	2 (15)	12 (42)
Tertiary	6 (46)	15 (52)
Did no respond	-	1 (3)

*** 2 PWSMA below age 18**

13 were PWSMA, comprising 8 females & 5 male

29 caregivers, comprising 18 females & 11 males

3.1.2 Psychosocial Impact of SMA

PWSMA and caregivers were asked about the psychological and social impact of SMA on them. Anxiety, stress and depression were the top three responses among both groups. Seventy-six percent of caregivers and 38% of PWSMA experienced anxiety. Slightly over half of caregivers (55%) and PWSMA (54%) reported stressful experiences, and 20% of caregivers and 38% of PWSMA reported having depression.

Full-time caregiving had caused 14% of caregivers to lose or give up their employment as the time commitment required for care and attending to the medical needs of the PWSMA made it difficult to keep regular employment. Three caregivers reported experiencing social isolation and two reported having had troubled relationships. PWSMA who were wheelchair-bound also experienced social isolation (31%) due to societal stigma or facilities that were not disabled-friendly (Table 2).

Table 2: Psychosocial Impact of SMA

Psychosocial Impact of SMA	PWSMA N = 13 (%)	Caregivers of PWSMA N = 29 (%)
Feel stressed	7 (54)	16 (55)
Feel depressed	5 (38)	6 (21)
Feel anxious	5 (38)	22 (76)
Feel socially isolated	4 (31)	3 (10)
Lost job	2 (15)	4 (14)
Experienced troubled relationships	3 (23)	2 (7)
Others	3 (23)	4 (14)
Did not respond	3 (23)	1 (3)

14% of caregivers lost or gave up their jobs

To further understand the psychological state of the participants, they were asked to fill out the DASS 21 questionnaire (Appendix 2). The results are shown in Table 3. Among the PWSMA, 15% scored moderately depressed, moderately anxious and severely anxious respectively. One (8%) PWSMA was mildly stressed.

The results from caregivers of PWSMA shows that 10% were mildly depressed, two (7%) were mildly anxious and one (3%) was mildly stressed.

Table 3: DASS 21 Scores for PWSMA and Caregivers

Dass 21 Scores	PWSMA N = 13 (%)			Caregivers of PWSMA N = 29 (%)		
	Depression	Anxiety	Stress	Depression	Anxiety	Stress
Normal	11 (85)	9 (70)	12 (92)	26 (90)	27 (93)	28 (97)
Mild	0	0	1 (8)	3 (10)	2 (7)	1 (3)
Moderate	2 (15)	2 (15)	0	0	0	0
Servere	0	2 (15)	0	0	0	0
Extremely severe	0	0	0	0	0	0

"Anxiety, stress and depression were the top three responses among PWSMA and caregivers."

3.1.3 Limitations on Daily Activities

The top three limitations felt by both PWSMA and caregivers were mobility independence, transferring and physical activities (Table 4).

Table 4: Daily Activities affected by SMA

Types of daily activities	PWSMA N = 13 (%)	Caregivers of PWSMA N = 29 (%)
Mobility independence	7 (54)	24 (83)
Transferring	7 (54)	17 (59)
Physical activies	7 (54)	16 (55)
Going to toilet oneself	6 (46)	15 (52)
Being independent	6 (46)	8 (27)
Turning in bed	5 (38)	11 (38)
Personal hygiene	4 (31)	15 (52)
Dressing oneself	4 (31)	11 (38)
Attending work/school	4 (31)	8 (28)
Social activies	4 (31)	6 (21)
Feeding oneself	3 (23)	5 (17)
Others	0	3 (10)

3.1.4 Symptoms Affecting Quality of Life

Caregivers and PWSMA were asked to identify the symptoms that currently have the most significant impact on their daily lives. Responses from the PWSMA revealed that the most significant symptoms that affect their daily lives were muscle weakness and joint contractures, followed by lung infection and fatigue. Meanwhile, the symptoms that caregivers of PWSMA regarded as having significant impact on the daily lives of their charge were muscle weakness and fatigue, followed by cough and joint contractures (Table 5).

Table 5: Symptoms affecting Quality of Life (QOL)

Symptoms Affecting QOL	PWSMA N = 13 (%)	Caregivers of PWSMA N = 29 (%)
Muscle weakness	11 (85)	24 (83)
Joint contractures	8 (62)	15 (52)
Fatigue	5 (38)	17 (59)
Lung infection	5 (38)	6 (21)
Cough	4 (31)	15 (52)
Breathing	4 (31)	10 (34)
Respiratory failure	3 (23)	6 (21)
Sleep	3 (23)	5 (17)
Falls	2 (15)	5 (17)
Communication	2 (15)	3 (10)
Bone fracture/hip dislocation	2 (15)	1 (3)
Feeding/swallowing	0	8 (28)
Paralysis	0	2 (7)
Others	0	1 (3)
Did not respond	1 (7)	1 (3)

3.1.5 Medical Care-Related Experiences

Participants were asked several questions related to their medical care experiences and expenses thus far (Table 6).

Among the PWSMA, nine did not require emergency visits or hospital admissions in the last 12 months, whereas four had sought emergency care or had been admitted to a hospital at least once or twice in the past year. More than half of the caregivers had brought their loved ones for emergency visits or hospital admissions in the last 12 months.

The majority of the PWSMA (62%) and caregivers (76%) had regular medical follow-up with varying frequencies of visits. PWSMA types 2 and 3, who typically survive childhood, and the caregivers' group (which included those who care for children with SMA type 1) have different supportive care needs and attended a number of specialist clinics/services. This is reflective of the wide range of multidisciplinary care needed to support PWSMA.

62% PWSMA & **76%** of caregivers had regular medical follow-up

Table 6: Medical Care-Related Experiences among PWSMA and Caregivers

Medical Care-Related experiences	PWSMA	Caregivers of PWSMA
Frequency of hospital admissions or emergency department visits in last 12 months		
None	9	16
1 - 2 times	4	10
3 - 10 times	0	3
More than 10 times	0	0
Frequency of hospital visits for routine follow up in last 12 months		
None	5	7
1 - 2 times	4	9
3 - 10 times	2	12
More than 10 times	2	1
Types of specialist clinics follow up in last 12 months		
Neurology	6	19
Physiotherapy	5	17
Respiratory medicine	5	10
Occupational therapy	3	9
Orthopedic	3	6
Complimentary medicine	3	6
Hydrotherapy	3	4
Genetic	1	10
Nutrition	1	2
Speech therapy	0	1
Mental health	0	1
Palliative care	0	0
Others	3	4
Did not respond	4	7
Types of medication prescribed or ever taken		
Albuterol (inhaled)	0	4
Albuterol (liquid)	0	0
Albuterol (tablet)	0	0
Carnitine	0	0
Creatine	0	0
Hydroxyurea	0	0
Steroids	1	1
Valproaic Acid	0	1
Sodium phenylbutyrate	0	1
Riluzole	0	0
Nusinersen/Spinraza	0	0
Others	5	7
Did not respond	7	14

Medical Care-Related experiences	PWSMA	Caregivers of PWSMA
Types of respiratory assistance used at home		
Cough assist device	3	7
Non-invasive ventilation	2	6
Chest physiotherapy	1	2
Postural drainage	0	0
High frequency chest wall oscillation	0	0
Suction to remove secretions	1	6
Invasive ventilation	0	0
Others	1	2
Did not respond	7	10
Ever undergone scoliosis surgery		
Yes	4	3
No	8	25
Did not respond	1	1
Estimated annual SMA related expenses (range in RM)		
Medical professional	N/A	2000-5000
Allied health	N/A	800-6000
Medicine	N/A	200-1000
Medicine supplies	200-360	2000-12000
Nutrition	1200	1000-6000
Orthotics	300	1000-5000
Home renovation	700-10000	2000-10000
Mobility devices	600-9800	1000-22000
Respiratory support	1200-1500	1200-20000
Travel & accommodation	300-1000	200-1200

Half of the PWSMA have visited neurologists and respiratory specialists, but only three have seen an orthopaedic specialist. Another three have sought help from complementary medicine practitioners, but none have ever seen a mental health professional.

In contrast, 19 caregivers have brought their loved ones to a neurologist and 10 each have visited a pulmonologist and geneticist respectively. Six caregivers have also brought their PWSMA to see an orthopaedic specialist and complementary medicine specialist. However, only one consulted a mental health professional.
Caregivers and PWSMA were asked about other types of interventions and therapies they have used to treat SMA symptoms. The majority has undergone physiotherapy, occupational therapy and hydrotherapy, but none have ever received palliative care support despite SMA being a life-limiting condition. At home, the three most commonly-used therapies for respiratory maintenance and clearance were a cough assist device, non-invasive ventilation and suction to remove secretions.

Around half of the caregivers (48%) and PWSMA (54%) did not respond to the question regarding medications and supplements that had ever been prescribed by a doctor for the PWSMA. Another five PWSMA and seven caregivers did not specify what other medications they were taking. The caregiver group reported that the most commonly-prescribed drug for their charge was inhaled albuterol (14%), followed by valproic acid, steroids and sodium phenylbutyrate. Almost a third (31%) of PWSMA had undergone scoliosis surgery.

3.1.6 Awareness on Current Drug Treatment Options

In light of the recent rapid advances and introduction of novel therapies for SMA, PWSMA and caregivers were asked several questions related to current drug treatment options. (Table 7). Almost 85% of PWSMA were aware of drugs developed to treat SMA, compared to 70% of caregivers. When asked to indicate what would be the most important outcome for a possible drug treatment, providing gains in function, such as increased strength and energy, and being able to do something the PWSMA was unable to do before, were the most important outcomes for both PWSMA and caregivers. In addition, 31% of caregivers chose the option of slowing or stopping disease progression (without improvements in QOL), although only one PWSMA chose this. No PWSMA selected the outcome of prolonging life on its own, although four caregivers (14%) indicated this as the most important outcome. Other PWSMA and caregivers chose lessening symptoms and improving QOL as the most important outcomes for a possible drug treatment.

When questioned regarding factors that would influence decisions to not use or stop a given treatment, the most common response in both groups was significant risks of serious side effects such as cardiac or kidney toxicity. Other common responses were the cost and the burden of administration (such as the need for anaesthesia, radiation exposure, surgical procedure, etc.), followed by the burden of treatment administration, such as the time that it would take away from daily activities, job, school, etc.

The majority of participants (85% of PWSMA and 65% of caregivers) have never participated in clinical trials largely because they were unaware of any clinical trial they could join. Other reasons given included trial enrolment being closed, additional burden (travel, potential risks, time missed from work, etc.) and having received access to an experimental drug via a compassionate use programme. Some of the caregivers' reasons were trial enrolment being closed or not getting any response to their enrolment query, and the additional burdens of joining a clinical trial.

The PWSMA and caregivers were then asked to select the most important factors influencing their decision regarding participation in a clinical trial of an experimental treatment. The top two responses were concerns about the risk of rare, but serious side effects and how the treatment might prevent further disease progression or improve one's health. Other factors selected were common side effects (headache, back pain and skin rashes) and how the trial might affect their current treatment plan.

Table 7: Awareness of Current Drug Treatment Options for SMA

Awareness on current drug treatment options	PWSMA	Caregivers of PWSMA
Aware of drug treatment for SMA		
Yes	11	20
No	2	7
Did not respond	0	2
Most important outcome for possible drug treatment		
To provide gains in function	9	10
To lessen symptoms	3	3
To stop or slow down disease progression	1	8
To prolong life span	0	4
Others	0	0
Did not respond	0	4
Factors influencing decision to not use or stop a given treatment (Can select more than 1 answer)		
Serious side effects	6	15
Cost	5	13
Time spent receiving treatment	5	4
Burden of treatment drug administration	4	9
Common side effects	3	1
Route of treatment administration	2	4
Duration of treatment	0	2
Others	0	2
Did not respond	2	5

Awareness on current drug treatment options	PWSMA	Caregivers of PWSMA
Ever participated in any SMA related clinical trial		
Yes	0	0
No	12	28
Did not respond	1	1
Reasons for never Participated in SMA related clinical trial before		
Not aware of any clinical trial	7	9
Tried to enrol but trial enrolment was closed	1	4
Burden of trial such as cost, and travels	1	2
Have received access to via a compassionate use program	1	0
Tried to enrol but no response	0	1
Unsure	0	1
Other reasons	0	3
Did not respond	0	3
Most important reason to participate in a clinical trial? (Can select more than 1 answer)		
The risk of rare but serious side effects	8	19
How it might prevent further disease progression	8	18
Common side effects	7	9
Reputation of study site doctor	3	7
Promise of receiving therapy on trial completion	3	3
Availability of safety data	3	2
How it might affect the current treatment plan	2	8
Duration of visits	2	2
Proximity of the study site	2	2
Frequency of visits	1	4
Did not respond	1	5

3.1.7 Awareness on Genetic Testing and Newborn Screening

Pre-implantation genetic diagnosis (PGD) of genetic disorders like SMA is a very efficient method of screening, especially for couples with a previously-affected child. PGD offers couples, who are carriers of genetic disorders, a realistic option of having an unaffected child without having to resort to prenatal diagnosis and therapeutic abortions. The majority of our participants have not heard about PGD. Only one (8%) PWSMA and eight (28%) caregivers have heard of PGD and would consider it in the future.

Prenatal genetic testing or prenatal diagnosis (PND) is available for couples who are carriers of SMA through chorionic villus sampling at 10 to 14 weeks of pregnancy or amniocentesis at 16 to 20 weeks of pregnancy to determine if the foetus has inherited two copies of the SMN1mutation. Couples who are or planning to get pregnant may access genetic counselling and consider the option to pursue PND. In contrast to PGD, slightly more than half of the caregivers have heard of PND and 40% of them would consider PND in the future. Ten percent have undergone PND and were very satisfied with the service. One had gone through it twice and had proceeded to termination of the affected foetus.

Newborn screening (NBS) identifies conditions that can affect a child's long-term health or survival. Early detection, diagnosis and intervention can prevent death or disability, and enable children to reach their full potential. Knowledge and advances in the genetics of SMA has allowed for the development of tailored therapeutic interventions, and when instituted early, has changed the clinical progression of the disease. However, the cost and access to such therapies remains a huge hurdle. Among the caregivers, 18 (62%) have heard of NBS and would consider it in the future.

8% of PWSMA and **28%** of caregivers have heard of PGD

Table 8: Awareness of Genetic Testing and Newborn Screening

Awareness on gentic testing and newborn screening	PWSMA	Caregivers of PWSMA
Head of Pre-implantation gentic diagnosis (PGD)		
Yes	1	8
No	12	18
Did not respond	0	3
Would consider PGD in future		
Yes	0	9
No	11	12
Did not respond	2	8
Heard of Prenatal Diagnosis (PND)		
Yes	3	14
No	9	10
Did not respond	1	5
Ever undergone PND		
Yes	0	3
No	11	23
Did not respond	3	3
Would consider PND in future		
Yes	4	12
No	4	7
Did not respond	5	10
Heard of newborn screening (NBS)		
Yes	5	18
No	2	5
Did not respond	6	6
Would consider NBS in future		
Yes	5	17
No	2	3
Did not respond	6	9

3.2

Qualitative Findings

The findings from the qualitative component of the study are described in detail below. It focuses on the experiences related to diagnosis, the impact SMA has on the lives of PWSMA or caregivers, as well as their worries and concerns. The participants' hopes and concerns for treatment, including recommendations to the Government, are also included.

3.2.1 Characteristics of Participants

Seven PWSMA, comprising four women and three men, were interviewed using IDI. Their ages ranged from 19 to 42 years old. Five of them have SMA type 2 and two have SMA type 3. All are currently residing in Kuala Lumpur and Selangor. Twenty-three caregivers, who are all parents of PWSMA, were interviewed through either IDIs or FGDs. The ages of their (surviving) children range from two to 23 years old. Seven parents went through the painful experience of losing their children with SMA before the age of one, and five lost theirs at age of three, eight and 16 respectively.

3.2.2 Experiences related to diagnosis

3.2.2.1 Experiences related to diagnosis - Developmental Delays

Most of the mothers noticed "something not right" with their SMA child either at around three months of age when their children were not lifting their heads, around four to five months of age when they were not turning, or when they were still not walking by the age of one year.

Mrs PP, mother of a SMA type 2 child who has passed away and a nurse by training, noticed that her child would not lift her legs at four months of age while her diapers were being changed, and had not lifted her head or turned over herself by the age of five months. She had a feeling that "something was not right".

Mrs SS, a mother whose younger child has SMA type 1, noticed that he still had not turned over by himself at four to five months of age and wondered if it had anything to do with an incident of him falling from the sofa at the age of three months.

Mrs KK who has five children, recalled how both she and her babysitter noticed that her youngest son was "different from others" from his birth up to five months of age. Her son is now a teenager and diagnosed with SMA type 2.

There were a few mothers who noticed certain signs as early as the first month of their child's life.

Mrs YY, whose child with SMA has passed away, shared her experience in an FGD:

> "At delivery he looked normal. He could move; his legs could also move until day 10. Ten days after that, his condition gradually got worse, like being lethargic up to a month. We went to the clinic for [vaccination] at one month and asked the doctor, 'Why is my child is so floppy? His hands were slow to react.' The doctor said, "Benda ini biasalah (This is normal)." Two to three days after that, I was still not satisfied. I went to the clinic; I went to see another doctor and he said my child was so floppy and felt like an old cloth. His neck was so weak and it was then that the doctor asked me to go to the hospital."

Mrs WW, whose child with SMA type 1 has passed away, related how as a then, first time mother, she noticed that her baby appeared normal during his first month of life, with the exception of his voice and cries being very weak.

Several other parents were alerted to their child's condition when their child was still not walking after their first birthday.

Mrs TT, shared how her older daughter - now in her 20s and diagnosed with SMA type 3 - had not walked even when she was over a year old and described her cultural belief on slowness in walking:

> "My first child - at the age of 10 months she could talk, but they usually say the one who talks first, walks later (yang cepat bercakap, dia lambat jalan). That is our culture, so okay. Then at one year old, she still didn't walk, was very slow. As I was working in a private hospital, my friends said that I didn't rest enough (mak tak kuat berehat) when I was pregnant, 'so don't worry, just go with the flow, she can walk later'."

Mrs LL, mother of a woman in her 20s with SMA type 2, was also concerned when her daughter was still not walking when she was over a year old.

3.2.2.2 Experiences related to diagnosis - Seeking A Diagnosis

Many caregivers described the experience of seeking advice about their child's delayed development and reaching a diagnosis of SMA as a long arduous journey filled with confusion and uncertainty. They often met dismissive, and even, insensitive health professionals, and experienced many misdiagnoses.

> "I think for about one year plus, we were sourcing for treatments - for one whole year! To our disappointment, we couldn't find anyone - nobody seemed to know. Many doctors do not know actually and this was 18 years ago, very sad to say. We also brought her for physiotherapy [and] the physiotherapist also didn't know about this illness."

Mrs LL, mother of a woman in her 20s with
SMA Type 2

Mrs SS, mother of a school-age boy with SMA type 1, had a similar experience of seeking advice from several paediatricians when her son was not turning over by himself by four to five months of age. She reported that the paediatricians told them that "No, nothing [to worry about] - everything is fine." Then when her son was six months old, she and her husband took him again to see two more paediatricians and was told that he could be delayed in his development:

> "So they said, 'Let's do some physiotherapy'. After six months of physiotherapy, he still wasn't showing any results. He would crawl a little bit, but he never really crawled by himself - we needed to assist him. And at one year old, I thought that he was not putting on weight - this whole time, he only put on 100 grams. So that doesn't make sense, you know, after all the food I fed him. I did a Google search. I asked why kids are not putting on weight and not crawling, and there was a whole list of disorders. I insisted the test be done. So we immediately did the test and it was confirmed to be SMA."

She went on to say that the doctor was unsure which SMA type he was and this added to their sense of uncertainty.

> "At that time, we were really lost. We didn't know what to do back then. After seeing a geneticist, he explained to us in more detail on what [SMA] means, and then added 'Nothing much we can do'."

The lack of knowledge or attention given to the delayed development of children by healthcare professionals is shown by what **Mrs WW**, who lost her first child with SMA type 1 a number of years ago, shared in a mothers' FGD.

> "But at least they have to know lah; they have to lah, I feel. Because they are medical people. Because the very first contact is at Klinik Kesihatan (Health Clinic) for the parents. So, for example, like my case ah, if my mother-in-law didn't tell me, then we wouldn't know about this. It was my mother-in-law who raised the question to the doctor, then only the doctor said "Eh, betul ha, macam tak sihat sikit (Eh, that's right, seems a bit unhealthy)." Then after that he gave a letter [of referral]. Because they don't know."

Mrs MM, who is a young mother, admitted that it was actually her son's babysitter who noticed the developmental delay and pushed her to get the boy checked when he was 16 months old.

> "The first doctor at a public hospital thought there was just a delay, [and said] 'it's nothing'. The babysitter persisted and so we took him to a private hospital. Doctors at the university hospital suspected SMA and did the test."

There were also parents who shared that they not only met with healthcare professionals who overlooked the delay in development, but also made insensitive remarks about their child. **Mrs UU**, mother of a school-age daughter with SMA type 2, shared how, during routine check-ups at the Klinik Kesihatan (Health Clinic), when she repeatedly alerted them to the fact that her daughter, then one-year-and-three-to-four-months-old, was still not walking, the response given was 'Eh, no lah, she is plump, she is lazy … there is nothing, no problem'. It was only when her daughter was admitted to the ICU for a fever when she was one-and-a-half-years-old that the doctor there noticed the delay and had her diagnosed.

Delayed diagnosis due to the poor awareness of healthcare professionals and delay in referral to a specialist was also the experience of **Mrs NN**, who has four children, two of whom had SMA type 1 and have since passed away. She shared that her second child's developmental delay was picked up by the Klinik Kesihatan (Health Clinic) doctor, who then referred her child to a visiting neurologist in Melaka Hospital. However, they had to wait three to four months for the appointment. Later, her child developed pneumonia and was seen by a private paediatrician, who referred the child directly to a paediatric neurologist in Kuala Lumpur Hospital. However, her daughter passed away at one year and four months of age. Her third child, who also had SMA type 1, passed away at 11 months of age

Mrs QQ, shared her own long journey of reaching a diagnosis for her son with SMA type 2:

> "After he was two, he fell sick and was diagnosed with pneumonia [and] got admitted at a university hospital. He was treated by [a paediatric neurologist], so after the doctor checked him and all, he said it looked like SMA, but that he wasn't sure. So he suggested to take blood [for the test], [but] if it's sent to IMR (Institute of Medical Research), then need to pay around RM700. I was okay with it even if needs to be paid. Then after that, he called me again and said his friend in university is doing research on SMA, so we shall send to him, no payment needed. So, we decided to send the blood there. We waited for almost a year and when I asked for the result, he said the file that contained the documents was lost, but via conversation, he said it's confirmed to be SMA type 2."

What is apparent from the above interview excerpts is that there is a lack of knowledge and very little attention or urgency given to developmental delay in infants among healthcare professionals, as evident from expressions such as "It's nothing" and "Biasalah (It's normal)".

"there is a lack of knowledge and very little attention or urgency given to developmental delay in infants among healthcare professionals, as evident from expressions such as "It's nothing" and "Biasalah (It's normal)"

The PWSMA, on the other hand, grew up knowing they are different from other children, but only got to know that it was SMA at a much older age, either through their parents or told directly by healthcare professionals. Most of them echoed the experiences of the caregivers, with their conditions being frequently misdiagnosed before finally reaching the diagnosis of SMA after many hospital visits. All were brought by their parents to healthcare professionals, except for one who sought help herself for her excruciating pain and was diagnosed with SMA when she was 19 years old.

> “I think growing up, it was kind of complicated because I always wondered why I was weaker than others. I thought there was something wrong. I didn’t really understand, like, for example, why couldn’t I run like the other boys? Why are they faster than me? Why do I get tired so fast? These are the things I always questioned, but didn’t understand until I was older and then I really knew that I have a condition called spinal muscular atrophy.”
>
> **Mr G, early 20s,**
> SMA Type 2

> “My parents didn’t accept the Western doctors’ diagnosis … When I was young, I was always brought up with the idea that I was lazy, because they always told me that you are lazy, that’s why you can’t lift your arms, you can’t walk, you can’t balance, if you fall down. They thought I was doing it on purpose. So I didn’t really have an idea what was happening when I was young … I just kind of lay on the floor, feeling negative the whole time; so, after I knew about the diagnosis, I started searching, learning and teaching myself about the disease.”
>
> **Mr A, early 20s,**
> SMA Type 3

3.2.2.3 Experiences related to diagnosis - Coping with the Diagnosis

Parents shared how they were devastated by the diagnosis of their child having SMA, particularly when they were told that their child would be short-lived and/or that there was no cure. They were in shock and disbelief when told that their child’s functional ability would deteriorate over time. Anger, frustration, hopelessness and sadness were the common feelings expressed during the interviews. Some of them were traumatised by the manner in which the doctor broke the news to them. Many of the participants, including fathers, were tearful while recounting their experience of receiving their child’s diagnosis of SMA.

Mr AA, whose youngest son has SMA, described the pain, devastation and shock he felt upon hearing his son’s prognosis:

> “At first, I did not know the seriousness of it, the doctor did not straightaway break the news to me. He just asked me to read [about it] and when I did, I still did not know the seriousness, but I did sense something was not too good. Bit by bit I faced the reality. When I realised the implications, it is very devastating, very painful to me and to my missus. At that time, I still had the hope that eventually there would be a chance of a cure … [although] I read about SMA and I knew that it’s a form of muscular atrophy, I did not know the seriousness [of it, and] had the hope of [a] cure, but the specialist told me my child needed to be confined to the wheelchair and that was a sudden shock to me.”

Mrs XX also noted her unreadiness to hear her two children’s prognosis, but admitted that having the information was an important step in understanding the condition.

> “At first, I was not prepared [to hear the news]. But thinking back, there were a lot of benefits to me. It was advantageous for me to know [about] my children’s condition.”

Mrs NN, who lost two children to SMA, recounted her shock and disbelief during and after receiving the diagnosis for her second child, the first of her two children who had SMA to be diagnosed.

> “I was shocked because he said genetic, meaning [that] I am a carrier [and] my husband is a carrier - like [it’s] passed down from generation to generation. But in my family, not even one person has SMA; [we’ve] also never heard anything about this disease. Was very shocked when the doctor said there is no medicine and no treatment. I didn’t think there was a disease that doesn’t have medicine and treatment. [And] my child looked normal, so I could not accept it when my child was nine months old and the doctor told me that my child was sick.”

“They were in shock and disbelief when told that their child’s functional ability would deteriorate over time. Anger, frustration, hopelessness and sadness were the common feelings expressed during the interviews.”

Mrs UU, mother of a school-age girl with SMA type 2, related how her husband is unable to accept the diagnosis, even now.

> "My husband, until now, he can't believe [our daughter] has SMA. He really cannot accept [it] - until now, he still keeps saying [that] he doesn't believe she has SMA. He said this must be because when I was pregnant with her, I had a fall. He keeps saying [that] during my pregnancy, I fell, and so [the condition] may be because of the stress (tekanan) because of my fall. I said, But we went through the blood test and all the check-ups, and confirmed she has SMA. 'No, itu tipu (it's a lie)', he said. So, up to now, he cannot accept it."

Anger mixed with disbelief is another emotion that came out in many of the interviews, particularly when told that their child would be short-lived. The quotes below from Mrs UU encapsulates the emotions:

> "Angry, I am really angry! I asked, 'Why?! It's not possible there is no cure! I don't believe [it]! How is it you have a disease that gets worse?'"

But for another mother, **Mrs XX**, who has two children with SMA, guilt was the primary emotion.

> "There is [the] feeling of guilt, like there was something wrong with my pregnancy, because I could not comprehend [it] at that time. At that time, we did not understand the extent of the diagnosis. But thinking back, after meeting with the doctor, getting counselling and all, [even] after one to two years, I [still] could not accept [the diagnosis]."

Mr CC, father of a man in his 20s with SMA type 2, said:

> "When we first heard the news, we were devastated. We were like zombies for a couple of days - like zombies, you know. And we had to console ourselves and … we picked ourselves up from there."

Mr DD, father of a son with SMA type 3, was in his words "terrified by the diagnosis".

Mrs KK, whose youngest son, now a teenager, was diagnosed with SMA type 2 at five months of age when he was brought to a geneticist at a university hospital, shared her frustration and sense of helplessness:

> "Felt frustrated and very sad as [it was] after four normal pregnancies. Feel lost as [I] did not have any knowledge about SMA. I was told that my son could live until five years old only. Explained that it was genetic, but none of my relatives and my husband's side have this condition. Then I read about SMA from the internet and could only find overseas support groups."

Mr BB who has four children, two of whom had SMA type 1 and who have passed away, shared that he was devastated by the way the doctor delivered the diagnosis of their first child when she was nine months old:

> "Doctors lack empathy and did not offer much help. The doctor said, 'Eh, you know or not your child ah, can't live long. I can only give you a period of a maximum of two years.'"

Perhaps the most heartwrenching experience was shared by **Mrs PP**, whose SMA child passed away at two years of age. In between sobs, **Mrs PP** recounted how she and her husband went through a very distressing experience when they were at the hospital waiting for the diagnosis.

> "[The doctor] asked us to sit. I couldn't sit, I couldn't be calm because I felt so cold, was shivering so much; my feet, my knees they all felt like … felt like my heart said yes. When the doctor said 'Your child does have SMA', what could I do? I was stumped, I was hugging [my baby]. Because I was hugging her, maybe that made me strong. I was numb, felt dizzy, felt like fainting. I said, Am I dreaming? Not only is this a disease I don't know about, then suddenly you say there's no cure? Okay, I thought not knowing the disease was okay, you can give me hope that there is some drug or operation or anything. When the doctor said there is no cure and added that she would have a short lifespan, maybe one or two years, for me, you don't need to tell me these things. I have cried until there are no more tears. Ah, my husband was crying, (I) couldn't look at his face. He was crying so I hugged my daughter. I was strong as long as she was there, I said in my heart, Never mind, accept it … accept it, it is the will of God."

3.2.2.4 Experiences related to diagnosis - Lack of information and supportive services

An overriding theme that emerged when participants related their experiences of reaching their diagnosis of SMA was the lack of information and supportive services. Most shared that in addition to the long journey of getting the right diagnosis, they also spent a lot of time researching for information on SMA, looking for support groups and seeking knowledge and skills on how to better care for their child with SMA. Others sought alternative and complementary treatments and cures, with some continuing to do so after the diagnosis.

"All the stuff that we know about SMA was through personal research. A lot of it was found out through Cure SMA, which is an American NGO, and Families of SMA.

"Throughout my life it has been hospital trip after hospital trip, strange acupuncture, Ayurveda - everything on the list pretty much - and then at one point, spiritual healers, which didn't mean anything because they'll be like. 'Oh, yeah, you have a ghost,' and I'm like 'No, you have a ghost.' It's like spinning the medical roulette wheel; I think the only thing I haven't tried is cupping."

Ms C, 32 early 30s
SMA Type 2

"We searched all about this disease. Then we searched for support groups. Then we found Mrs SS. So, we were motivated … from the group, we could see many SMA patients - all have successful stories - then we became motivated to take care of our son."

Mr AA, father of a young boy with SMA Type 3

"…in addition to the long journey of getting the right diagnosis, they also spent a lot of time researching for information on SMA, looking for support groups and seeking knowledge and skills on how to better care for their child with SMA."

3.2.3 Impact of SMA on Patients and Families/Caregivers

Among the PWSMA, the key themes that emerged were self-doubt; confusion over being special, different or inferior; a quest for independence; acceptance of additional challenges due to their deteriorating functional abilities; and frustration and depression as a consequence of abuse.

3.2.3.1 Impact of SMA on Patients and Families/Caregivers - Self-Doubt and Inner Turmoil

"Around the age of seven or eight, I realised that I was always going to hospitals for check-ups and this and that. Then I talked to my parents and they told me I am not like other kids. At one point, I felt really, umm, my friends calling me special, it sounds like … what am I? Am I special? Or I am not special (laughs)? Yeah, I was stuck between these questions, and then I just accepted that I am actually unique, I am different from the rest."

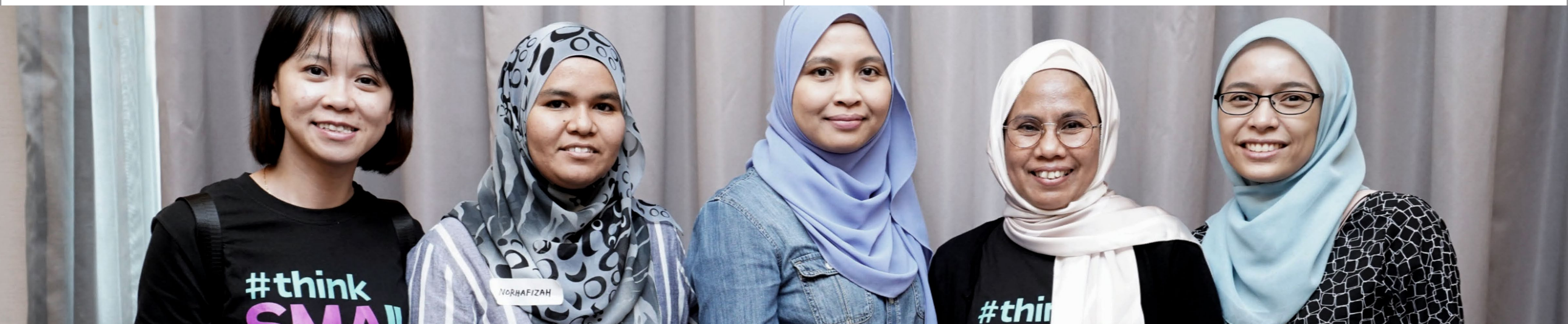
Mr G, early 20s,
SMA Type 2

"There was always physical abuse, emotional abuse, isolation, emotional blackmail. I compared myself to my brother and saw that he was being treated normally while I was ill-treated."

Ms A, early 20s,
SMA Type 3

"Sometime in my twenties, I needed others to assist in showering me. During my childhood years, I was able to bathe by myself. Back then, I still felt quite okay. Similarly, previously, during the night hours, I did not need to trouble others, asking for their help. But as I grew older, every night during the sleeping hours, I needed to trouble others to help turn me from one side to another. So I feel that almost daily, 24 hours, I need to depend on others to assist me. I feel that I am a very troublesome person."

Ms D, early 40s,
SMA Type 2



3.2.3.2 Impact of SMA on Patients and Families/Caregivers - Social Life

All except one PWSMA enjoyed the support of their family, including extended family for some. Two women, one with SMA type 2 and another with type 3, are living independently, with the support of personal assistants in activities of daily living. All have some friends and participate in social activities, but face challenges and limitations imposed by concerned parents, inaccessibility of public places and lack of supportive infrastructure. Similarly, their ability to have personal and financial independence is hampered by the general lack of barrier-free and disabled-friendly environments, and compounded by their deteriorating functional abilities, including, for some, respiratory problems.

“All the PWSMA have some friends and social activities, but face challenges and limitations imposed by concerned parents, inaccessibility of public places and lack of supportive infrastructure.”

“This condition actually makes me closer to my family, because I rely on them for help. And by relying on them for help, I actually form a good bond, a good relationship, whether I like it or not (laughs). I think, without [SMA], I will probably be on my own, you know, probably will not be spending much time with them.

“Due to my disability, I can’t play sports and all, so I always sat in the corner and watch the other kids play football. So I always felt left out, but I tried to fill up that gap by … I guess by talking. Yeah, that’s something we can do, talking about things we love, movies we love.”

Mr G, early 20s,
SMA Type 2

“I am very well supported by my very positive-thinking parents, but other family members, they just see that my sister and I can’t walk, can’t do things. Even the well-educated ones, for example, a cousin who is a doctor, when we tried to explain about SMA, they don’t really want to know.”

Ms B, early 20s,
SMA Type 3

“Hang out every other day. Ah … amazing people. They are all either extremely creative or extremely intelligent. And then we do whole dinner-table conversations, outside with like usually six to a lot of people.”

Ms C, early 30s,
SMA Type 2

“Yes, I do have a group of close friends. I knew them from when I came up to KL till now. We communicate through phone calls; sometimes, we have gatherings. Whenever they are available, they will drive and fetch me from where I stay. They will carry me into their cars and we will go out for a chat. Some of them still stay at the handicap centre. Some of them are my former colleagues too.”

Ms D, early 40s,
SMA Type 2

3.2.3.3 Impact of SMA on Patients and Families/Caregivers - Acceptance & Coping Emotionally

Among the caregivers, the key themes that appeared were struggles in acceptance, and coping with stress, anxiety and burnout.

“We have come to accept it. A lot of people see the way we provide for my child and ask if I am tired or not. And I say yes, it is definitely tiring. They ask if I am angry. I say I am not angry at all.”

Mr AA, Father of a 21-year-old man with
SMA Type 3

“It doesn’t matter since there is no treatment. No matter how, they are still your child, so…”

Mrs MM, Mother of a young boy with
SMA Type 3

“Of course we accept - it’s our religious view, it’s God’s will. So, my wife and I believe that we have to accept this with an open mind.”

Mr DD, Father of a preteen boy with
SMA Type 3

"I was so focused on my child that I would never, not even once, turn away from her in the room. Also, after she has finished feeding, I would just sit in the room. I did not cook, I did not go anywhere, because I was scared that she would have difficulty breathing. After having two children with SMA, I became paranoid. I became very scared. I had like a phobia too, as every time my child choked, I was scared. My hands shivered."

Mrs NN, Mother of two deceased children with
SMA Type 1

"My wife and I gave 100% to him. We sat at home only. We didn't bring him out. We get worried if we bring him out - worried that it would be easy for him to get an infection."

Mr BB, Father of two deceased children with
SMA Type 1

"I work. My hubby is also working, but men are tougher. Women, when we returns home after work, we will be stressed - stressed from work and bring it home. [Then] at home, I will have to handle [the chores]. Sometimes, I feel burnt out."

Mrs XX, Mother of two children with
SMA

3.2.3.4 Impact of SMA on Patients and Families/Caregivers - Changing Lifestyles

Caregivers had to cope with a rapidly changing lifestyle, with many becoming more responsible and patient, with a stronger belief in their faith.

"I tend to cut down on all my social activities. So family is everything. We take him as much as possible for family outings. Furthermore, going out as a couple has become a rare occasion, as at least one parent has to stay at home in the evening."

Mr CC, Father of a man in his early 20s with
SMA Type 2

"Our lifestyle changed. For me, I like sports, but I cannot do that now because my child cannot play sports. I like jungle trekking and camping, but we cannot do that. But if he wants to go to the beach, we go to some resort that is on top of the water."

Mr DD, Father of a preteen boy with
SMA Type 2

"For family occasions, kenduri, weddings, I would reject invitations from all my relatives; I never went for any."

Mrs PP, Mother of a deceased child with
SMA Type 2

3.2.3.5 Impact of SMA on Patients and Families/Caregivers - Within the Family

Impact of SMA on Patients and Families/Caregivers - Within the Family (Husbands and wives)

In FGDs with mothers, all shared that they are thankful to have very capable and supportive husbands. Others revealed some of the tensions that inevitably arise between couples going through the journey of confronting the disability of their child(ren), and for some, the death of their child(ren).

"At times we became angry. Then we calmed down. The test from God is great."

Mr HH, Father of two surviving children with
SMA

"I have no issues with the relationship with my wife, because both of us have already come to understand [the situation]. Between husband and wife, both of us, nothing is changed, we are still the same, our relationship is still the same."

Mr CC, Father of a man in his early 20s with
SMA Type 2

Impact of SMA on Patients and Families/Caregivers - Within the Family (Balancing care of other children)

The care of a child with SMA means that any other children (their siblings) are given less attention, and therefore, often felt neglected. **Mrs SS** shared the experience of how her older child started hurting himself when he was four-years-old, in order to get her and her husband's attention. He was referred to see a child psychologist.

Parents were aware of how their other children might feel neglected, as shown by the words of two fathers below:

"Sometimes we can't take care of our daughter, because we are paying more attention to our son and we do feel bad."

Mr CC, Father of a man in his early 20s with
SMA Type 2

"The other children, when they were very young, I could sense that they may be a bit resentful because of the attention given to the young one, but as they grew older, I explained [the situation] to them frequently and they came to understand over the years, became more accepting of their brother's situation and learnt how to maximise his happiness and his comfort."

Mr AA, Father of a 21-year-old man with
SMA Type 3

"The care of a child with SMA means that any other children (their siblings) are given less attention, and therefore, often felt neglected"

However, in another family where the child with SMA is the youngest, his older siblings took turns to help care for him.

> "His siblings are all very helpful. Even when [my youngest son] was younger … they took turns to help bathe him, change his diapers …"

Mrs KK, Mother of a teenage boy with
SMA Type 2

Impact of SMA on Patients and Families/Caregivers - Within the Family (Challenges of Parenting the Child with SMA)

The challenges of parenting are compounded when a child has SMA, as the parents are in dilemma over how they should handle certain situations.

> "So, his temper is getting worse. At first, we felt pity for him, so we gave in to him. But now I think we need to readjust - this is something that I really have to do."

Mrs MM, Mother of a young boy with
SMA Type 3

3.2.3.6 Impact of SMA on Patients and Families/Caregivers - Financial Strain

While sharing about the impact of SMA on their lives, participants also raised the issue of financial strain on the family. One cause of increased expenditure was the need to modify home infrastructure, such as renovating toilets to accommodate wheelchairs. Another was due to the fact that family outings with PWSMA need to be to places with disabled-friendly facilities, which can incur higher expenses. Most of all, necessary supportive equipment such as ventilators, are costly and not covered by medical insurance.

> "From the aspect of cost, we bought the oxygen machine, suction machine and all that on our own. We did not receive any help from anywhere. At that time, my husband was still in the public sector, but nobody told us that these things could be claimed. The hospital only told us to get it. … So my husband looked [for the machines] and we took out our savings. Everything we found on our own. We spent altogether RM30,000 and medical insurance did not cover [it]."

Mrs NN, Mother of two deceased children with
SMA Type 1

"Most of all, necessary supportive equipment such as ventilators, are costly and not covered by medical insurance"

3.2.3.7 Impact of SMA on Patients and Families/Caregivers - Education

The experience of schooling for PWSMA varied from two who had a very supportive school environment to the more common experience of an uncaring, unsupportive environment and episodes of being isolated and bullied. One common experience had by both PWSMA and caregivers is the assumption on the part of education officials that a child with a physical disability should be placed in the special education class (i.e. due to learning disability), even though the risk of learning disabilities for PWSMA is the same as the general population. Parents also lamented the negative experiences of their child being rejected from mainstream schools. Several participants share their experiences below.

Mr G: Uncaring environment and being bullied. I think that primary school was the hardest

> I think that primary school was the hardest time of my life because I faced many challenges. For instance, my classroom was on the first floor and the teachers weren't very … I would say, supportive. My parents talked to them, but they still didn't put my class downstairs, so I had to climb up the stairs. Sometimes I was so tired and my dad had to carry me up.
>
> And then there was the library, which was on the third floor (laughs). I remember all these bad memories: sometimes, there would be an activity in the library, and by the time I climb up the stairs back then I could climb up the stairs, but it was very slow so by time I climbed up the stairs and reached the library, the kids were coming out and it was time for the next class!
>
> And it's crazy, you know, the teachers didn't even care! Sometimes, I would have to rely on my friends to carry my bag for me. So if everyone is gone, I'm stuck I would have to wait for a stranger to come and help me.'
>
> In primary school, I also got bullied a lot. I was walking back then, but I walked in funny manner I would swing to the left and the right. I think that schools should be a safe environment, where students can have fun, and yeah, all this bullying shouldn't be there, shouldn't be tolerated…

Mr E, now in his late 20s, started in a special education class in a national school. He was in the special education class for two years before the teachers realised that he is of normal intelligence and moved him to a regular Year Two class.

> "Technically, I was late one year. Because they thought, besides the physical challenge [that] I was also mentally challenged. I don't know whether it was the school authorities or (pause) someone from the Government."

He recounted the sole incident when he was bullied:

> "There was one incident (long silence) of bullying because … because of my condition, I couldn't lift my head up. If I fall down now, I can't lift my head up. Yeah, so … my classmates when I was in primary school in normal class … one of them pushed my head down … and … and pressed. Well, of course, as my head was down, I couldn't lift it up (silence). Yeah … During those days, my helper would wait for me outside the class. So after a few moments, one of my classmates called my helper and she came in to help me and lift my head up. That was the only incident."

Ms B, who has SMA type 3, recalls that her school and teachers were not accommodating of her situation, but that was offset by her helpful friends.

> "Primary school and friends were fine, but the problem was the teachers, they wanted to put me in a class on the second floor and my parents had an argument with them … From Year Four onwards, my class was on the second floor; my dad had to carry me, but my friends (both girls and boys) were very good and helpful - they would carry me together with my wheelchair."

The following accounts from parents show how the attitudes and practices of schools and education officials can make a difference. **Mrs LL**, whose SMA type 2 daughter studied in a national Chinese primary school in Perak, shares her positive experience:

> "It was easy - the primary school, the teachers, the school were very understanding … They actually even had seated toilets [and] they specially made one OKU toilet. Normally, these primary schools don't have [these toilets]. Maybe when she was in Year Two, they applied to get the OKU toilet, so I think it was easier for my daughter."

However, the account from **Mrs TT**, whose daughter has SMA type 3, illustrated that schools in general are unwilling to accommodate and support students with disabilities - in this case, those with SMA.

> "I decided to send my girl to a private girls' school. I thought a girls' school would be more protective. A month later, an incident happened that caused the deputy principal to complain [to me] that my daughter's teacher had complained. It was because I requested the teacher to let [my daughter] sit at the front, and the teacher had said okay. I also requested for her to be allowed to use the teachers' toilet as it was nearby, because [she] couldn't walk too much at that time. She would struggle to walk and need to hold on the railing, so I requested the teacher to let [her] have her recess in the classroom. I even met the security guard of the school to help me monitor [her], and as my office was just behind the school, to give me a call if there was any problem.
>
> "Somehow, [my daughter] had an accident, she fell down in class and in the toilet, but no one called me. [She] told me when I came home in the evening. She persuaded me not to get angry and said it was not the teacher's fault, but hers. So, I said, Okay, it's fine. Once I reached home from school, the teacher called me. She said they couldn't have [my daughter] unless I have someone or a full-time PA (personal assistant) look after her. The teacher reasoned that if she assists [my daughter], the class would stop midway, disrupting it. The teacher even mentioned that the PIBG had complained that it would interrupt the class if [my daughter] continues to be there.
> "I was shocked! So shocked! I felt like it was not fair for my kid, but if I let [her] stay in that school, I was afraid they might do something to my daughter, so I took her out from that school immediately.
>
> "After all the commotion, I wrote a complaint letter to SUHAKAM (Human Rights Commission of Malaysia). I consulted a few friends, and to my mind, because it is not fair for me and the other kids, I needed to teach them a lesson. So I wrote a letter to the Ministry of Education and they sent officers to the school to investigate. After the investigation, they called us for a round-table meeting with my husband, SUHAKAM's commissioner and the school principal, but that deputy principal didn't turn up. It was another deputy principal that came. The teacher didn't want to confront me. The meeting lasted two hours and I brought up everything. According to SUHAKAM, the teacher was transferred out. Malaysia wasn't disabled-friendly those days - we were far behind, so that was my challenge!"

3.2.4 Worries and Concerns

When discussing worries and concerns, independence was a recurring theme. This is in terms of functional ability and financial capability, and closely related to issue of continuing care by family members or paid caregivers. The ability to lead a normal life is particularly imperative for parents with adult SMA children and the PWSMA themselves.

Participants also shared their disappointment, loss of motivation, helplessness, and anxiety and uncertainty for their/ their children's future, with one PWSMA disclosing that she has bouts of depression. Social discomfiture and community acceptance were also topics shared across all IDIs and FGDs.

The participants were mostly open about the topic of premature death and dying, and the need to make difficult treatment choices and advanced care plans was also mentioned in some IDIs and FGDs.
The participants were also given the option to indicate the level of each of their worries and concerns on a scale of 1 to 10, and the results are presented here where relevant.

3.2.4.1 Worries and Concerns - Independence

Worries about independence from both functional and financial points of view are top of the list for both PWSMA and caregivers. Independence is dependent on a number of factors: financial, infrastructure and social support. It is about having the financial means to engage a personal assistant who can attend to their daily physical needs.

Mr G, who has SMA type 2 and is completing his degree at a local university, rates concern for future independence eight out of 10. He said:

> "I worry the most right now about not being able to live independently in the future. Yes, because I know my parents will not always be there for me, so am wondering if I will be able to find a way to live independently. You know, have a good income, have someone to take care of me and be like everyone else when my parents are not there. So that's what worries me the most."

Mr FF, father of a woman in her early 20s with SMA type 3, has the same worry:

> "How is (my daughter) going to survive without us? That is really the greatest worry. So far, now I can still support her. When we are not around, she needs to self-earn. What can she do? That will eventually come to her."

Mrs LL, mother of woman in her early 20s with SMA type 2, was the most worried (with a rating of 10 out of 10) about having a reliable and dedicated caregiver to take over the care of her daughter when she herself is no longer able to:

> "And of course, the second issue is that I'm getting old and she is getting bigger - what if one day I'm not able to take care of her? If she stays at home, if someone can take care of her, then at least this person can replace my role if I am not around one day. Because she is a girl, right? My husband won't be able to take care of her bathing and all that."

Mr F, in his late teens with SMA type 2, is still fearful despite having siblings who have been caring for him:

> "I am scared [for] when my mother and father are no longer here. I would have to think about who else can take care of me. My siblings all have their own families."

For PWSMA who either do not have family support or who choose to live outside their family homes, the worries about independence were very much tied to the financial ability to engage a personal care assistant, as well as the availability of such an assistant. The excerpt below reflects the worry and concern of the two women who live apart from their family:

> "This is my biggest concern, because if I can't find a helper, I would feel like I have lost both limbs and not be able to function."

Ms D, early 40s,
SMA Type 2

3.2.4.2 Worries and Concerns - Fear of Losing Functional Abilities

The worry about independence is very much related to the reality of their deteriorating functional abilities. For **Mr G**, in his early 20s with SMA type 2, he was very conscious that the progressive weakness can happen very quickly and rated it seven out of 10.

> "Because with SMA, you get weaker with time - progressive weakness - I worry about losing the ability to do things I like and the things that matter, for example, just being able to hold the fork and spoon. … It is something basic, but I am losing my ability to do it, so I worry about all this.

Of even more concern is the deterioration of the respiratory system, leading to reliance on breathing apparatus, as mentioned by **Ms D**, who has SMA type 2:

> "My most worrisome situation is when I suddenly have phlegm in my lungs [and] I cannot cough it out and the helper is not able to push or rub my body correctly to expel it. Another major concern I can think of maybe is deterioration in respiratory function. I may not be able to take it as I am scared and also worry too much over this declining respiratory function. I am concerned that when the time comes, I may need to rely upon and be dependent on a respirator when my respiratory system deteriorates."

Mr E: An account of his daily struggle and coping

> Cough assist machine (CAM) has often enabled me to do my daily activities without being distracted by phlegm. Before I had started to use CAM, I was frequently struggling with phlegm in the morning as I always wake up with phlegm in lungs. Occasional coughs would affect my attention in the work I was doing like reading and writing.
>
> Both reading and writing are the main activities in my work in legal education or in the area of law generally. Although the twin activities can be considered as physically passive, they require deep concentration in my train of thoughts, especially when I work on academic pieces. So, before I had CAM, it was tiring for me to do well.
>
> Whenever my lungs were congested with phlegm, my carer had to press my chest to assist me to cough out the phlegm. Not only would this derail me from what I was doing, it would also sometimes cause chest sores.
>
> Since I began to use CAM, I have been able to cough out my phlegm in the morning. It has assisted me to clear my lungs before I start the day. It has also made coughing out better as my carer need not press my chest as hard and frequent as before. When she does, I also find it easier to cough out my phlegm. All in all, CAM has improved my lifestyle.

3.2.4.3 Worries and Concerns - Leading a Normal Life

Young PWSMA shared that they would like to be able to live a regular life like their healthy peers.
Ms B, in her early 20s with SMA type 3 and whose younger sister also had SMA and passed away said:

> "Maybe if I'm given a chance, I can be independent, because my parents right now are a bit overprotective … maybe because of my sister. I try to negotiate, like when going out with friends, we go on the MRT, but I don't [need to] inform my mum or both my parents. It's like slowly being released from being overprotected. "I wanted to go out to find a job, but I know my condition - it doesn't allow me to stay long hours in office - so I turned down a few job offers and am sticking to doing what I am capable of."

For parents whose children are young adults, the desire for them to have a normal life is conflicted with the reality of their situation and their desire to protect their child. **Mrs TT**, who has a daughter in her early 20s with SMA type 3, said:

> "Of course, I want her to get a partner too, to go on outings, right or not? That's a normal feeling, right? But then, can I let her go just like that? Can I cari (find) a boyfriend for her? Can I? I know love has no boundary, but can I do all that?"

The wish and the worry of whether they can have a "normal" romantic relationship is reflected in what **Mr E**, in his late 20s said:

> "Actually, what I am most worried … is [silence] is whether I'll be able to start a family"

3.2.4.4 Worries and Concerns - Limitations on social activities

Leading a normal life and limitations on social activities are two sides of the same coin. The latter is very much tied to accessibility of transport, infrastructure and public amenities. **Ms A** in her early 20s with SMA type 3, lamented:

> "I don't have a vehicle, I don't know how to drive, I can't drive, even though I want to. The infrastructure in Malaysia is really incomplete, because you have these disability pathways that suddenly break off in the middle and you don't know where they lead to … "A huge problem with the lifts … if people are trying to get in the lift, even though they see there is a [person in a] wheelchair there, they don't wait for me [to go in]. So I just drag myself to the side, and I just let them all go in, and I will just wait for the next one … "The facilities are not maintained [and] the inconsideration of people parking in the disabled spot."

Ms C, in her early 30s , who has SMA type 2 and uses a wheelchair, shared the inconvenience of using public transport:

> "The MRT is not very accessible. Last month, there was a problem for me trying to get to the MRT using [the] Grab [e-hailing service] or taking a bus. To take a bus, I have to go to the bus stop with pavements that don't work. So, I will be fighting on the road. I have used Grab before, but it is difficult for the driver to fold the wheelchair and communication is another problem."

3.2.4.5 Worries and Concerns - Gaining Employment

Parents with SMA children who are young adults and either completing or have completed their studies at tertiary level expressed concern if they were able to gain employment due to inaccessibility of workplaces.

For example, **Mrs LL** worried if her daughter in her early 20s, who is currently pursuing an undergraduate degree, will be able to secure employment suited to her qualifications. Her hope of her daughter being able to work from home was met with a disappointing response from a big company that claimed the private and confidential nature of the job did not allow staff to work from home.

> "In Malaysia, a lot of places are still not accessible to wheelchairs - this is one of the concerns. Even if we want to bring her to as many places as we wish, because wheelchair accessibility is very poor in Malaysia, sometimes we won't bring her there. We can't, so we don't go because of the inconvenience."

The lack of support and disabled-friendly infrastructure in Malaysia that limits the opportunities for PWSMA to be in the workforce is also lamented by **Mr CC**, whose son is about to graduate soon.

> "From the point of view of the infrastructure, public facilities like public toilets are not disabled-friendly. The whole list can go on and on whichever place you go. And of course, the working place - if he starts working, we of course would expect that environment to be disabled-friendly."

3.2.4.6 Worries and Concerns - Financial Expenses

The cost of raising a child with SMA is exorbitant, beginning from testing and treatment to the necessary purchase of medical devices, mobility aids, ventilators, BiPAP, CPAP and a cough assist machine. All are items beyond the reach of most families. Parents are also worried about the cost of treatment, surgery and assistive devices that help improve declining functional abilities. On top of that, treatment and aids for SMA are not covered by insurance, forcing some parents to take up a loan or source for charitable assistance in order to be able to obtain such treatment and items.

The experience of **Mrs PP**, whose daughter had SMA type 1 and has passed away, provides a clear illustration of the potential pressure on family finances. In her case, she and her husband were told that they needed to purchase a CPAP for their child:

> “Well, I didn’t have any money … I had been on half-paid leave for the past six months to a year or so. So I pleaded with the doctor, Help me arrange it with JPA. The doctor found out that with JPA, you need to pay first. He said, You have to pay first. I asked, How much is the quotation? He said, Around RM5,000. I said, Oh no, I don’t have the cash. What am I going to do? He said that he could help provide a referral letter [for us to apply for financial aid]. In the end, I received [a donation of] RM1,500 from the Majlis Agama Islam Melaka (Islamic Council of Melaka). Eventually, my colleague was able to arrange for me to get the CPAP machine on a loan, paid in instalments, and later [we] ended up getting a discount.”

Some parents from the lower income group need to take up a second job in order for their family to survive.

“My husband has two jobs. One, the ‘real’ work, then every evening, [he has] part-time work.”

Mrs XX, mother of two children with SMA

“Because I work as a lorry driver [and my wife is not working], our income is based on the trips made. If I don’t work, then there is no income - no monthly basic salary. I worry too if I don’t work. During the time my son was admitted into hospital. I had to take leave for one week [and couldn’t make any money].”

Mr EE, father of a baby boy with SMA Type 1

“Parents are also worried about the cost of treatment, surgery and assistive devices that help improve declining functional abilities.”

3.2.4.7 Worries and Concerns - Mental and Emotional Well-Being

Both PWSMA and caregivers frequently experience a wide range of negative emotions, including stress, anxiety, hopelessness, frustration, uncertainty and helplessness, due to their current situation and concern for the future.

Worries and Concerns - Mental and Emotional Well-Being (Loss of Sleep and Stress)

Mrs NN and **Mrs RR** both had a pair of children who succumbed to SMA type 1. The two mothers shared how caring for their children affected their sleep.

“Ah, at night, I never slept soundly. Every once in a while, I would get up to pray. I would recite [prayers] with my husband. I sleep, I recite, we look at her - we look at her every night. I can’t sleep because I am scared [that] when I wake up, she won’t be here because … Every night I can’t sleep (voice chokes with emotion). “With [child] number three, I was even more scared. Because with [child] number two, I was at the hospital, so I thought if anything happened, the doctor is here, the nurse is here - I can call them.”

Mrs NN, mother of two deceased children with SMA Type 1

“My family stays far away from me. My husband works in shifts. He only comes back at night for a week (In a month? Or in how long?). If he is on the night shift, I will be left alone with my child. I can’t sleep if my husband works till late night; I always check on my child to see if she’s breathing well or not, because I am very scared. No one is here with me.”

Mrs RR, mother of two deceased children with SMA Type 1

Worries and Concerns - Mental and Emotional Well-Being (Frustration and Hopelessness)

Two PWSMA expressed their frustration and despair at their condition:

“I lost motivation when I found out that no matter what you do, you can’t get strong, you can only maintain” At one point, it really got tiring just to maintain. I remember there was one time I was so motivated, you know. I signed up for a membership in the gym and all. I was going to the gym, and then, eventually, it just dies out, because even though I go to the gym, when I come home, I feel much more weaker. It’s just that I felt it was already pointless, and that’s how I lost motivation and I stopped doing physiotherapy.”

Mr G, early 20s, SMA Type 2

“I feel [frustration and hopelessness] most of the time, I don’t know why. Maybe because I am alone, so I get overwhelmed with everything. Maybe it is too much to bear sometimes … I also have my limits, sometimes I keep pushing myself and I am like: AAHHH! And just, you know, spiraling into despair.”

Ms A, early 20s, SMA Type 3

Worries and Concerns - Mental and Emotional Well-Being (Uncertainty and Anxiety)

Mrs MM, whose younger child has SMA type 3, is worried whether her other sons are also affected by SMA:

> "But anyhow, I haven't done a genetic test on my older children yet, so I'm not sure. I don't know whether they're SMA kids as well, because my younger son is type 3 with four SMN2 gene copies, so is considered a strong type 3. But a strong type 3 means that they can, you know, start to show symptoms at the age of 10 or 13. My kids are just eight and five, so that is the only concern that I have (slightly teary). Hmmm, maybe it's over-worry lah. My husband says that I always worry. But when they suddenly fall down, I will be like ... Oh! (gestures to show anxiety)"

Uncertainty and anxiety also surround young parents who are still thinking about having more children, as they have to consider the possibility that their future children could have SMA too.

> "I know this is genetic and genetic problems can affect future pregnancies."
>
> **Mrs XX, mother of two children with**
> SMA Type 1

Worries and Concerns - Mental and Emotional Well-Being (Depression and Loneliness)

One PWSMA shared how she can slip into depression, especially with the lack of support from her family and rated her worry for her own mental well-being as a 10 out of 10:

> "I have a lot of things to worry about because I don't have support from my family. Most of the time I am alone, especially in dealing with problems - just by myself, no one to turn to - so it can be quite difficult. Sometimes I feel really tired and the suicidal thoughts would come again and repeat themselves. Am trying to find a solution to overcome the problems that I face."
>
> **Ms A, early 20s,**
> SMA Type 3

Worries and Concerns - Mental and Emotional Well-Being (Grief and Sadness)

The grief of caregivers who have lost their child(ren) was expressed directly or indirectly while sharing the impact of SMA on their lives and when discussing their worries and concerns. The death of a child, although in the past, can still impact a family's life, especially when there is an anniversary or when they visit a certain place that reminds them of their lost one.

Mrs YY had just lost her child months before the FGD she attended. She tearfully shared how often she still remembers and grieves for him:

> "When I entered here (the hospital that was the venue of the FGD), I remembered. When I reached here, I remembered the time I was with him (sobs). Also, going to his grave; every Friday, I'll go. I feel like it was just yesterday that he left us ... like I didn't get enough time with him. I remember the time after Maghrib (dusk); he always cried because he was uncomfortable. I remember when I cuddled him, I hugged him, at that time. Therefore, at this certain time, at this specific time, the memory comes back."

Meanwhile, **Mrs WW** shared tearfully how hurt she felt by some of the things people would say to her about the death of her baby boy years ago.

> "Some people commented, 'Don't worry, you're still young, you can get pregnant again.' It's very hurtful, you know?! No one can replace anyone. Even if he is no longer here, even if I have 10 babies, 10 more kids, we cannot replace this baby. Even now, with our second child, we talk, we tell him, you had a big brother before."

Ms B, who has SMA type 3 and who lost her younger sister to SMA, shared:

> "When my sister was alive, we used to attend hydrotherapy at WQ park. But I stopped going after my sister passed away. My mother doesn't want to go there again, because she said that the place reminds her of my sister."

"... Sometimes I feel really tired and the suicidal thoughts would come again and repeat themselves."

3.2.4.8 Worries and Concerns - Premature Death

In most of the FGDs, the participants themselves brought up the issue of confronting premature death as one of their top worries and concerns. In the other FGDs, this topic was introduced by the interviewer. A range of responses was shared, from intense fear about losing their child to "not thinking about it" and facing death squarely.

Worries and Concerns - Premature Death (Fear of Death)

"I was just waiting for the time. The doctor had already told me that his lifespan is around two years. So, because of that, I was even more scared as the months went by."

Mrs NN, mother of two deceased children with
SMA Type 1

"Another thing that concerns me the most is sleeping at night. I feel like I might not wake up from sleep."

Ms D, early 40s,
SMA Type 2

Worries and Concerns - Premature Death (Being Prepared)

"So, the worry is there, but I don't want to worry too much. I'll burn out."

Mr CC, father of a man in his early 20s with
SMA Type 2

"Of course, everybody will die, right? Those with good health can also die at any time. What I mean is, for our case, we must give our best in order to avoid that lah. But if that's the fate, we accept it."

Mr DD, father to a preteen boy with
SMA Type 3

"His body shape, worsening lung function and when he could not breathe … I told everyone [in the family] to prepare for it, but have never discussed this with [her son] or my husband."

Mrs KK, mother of a teenage boy with
SMA Type 2

"I think about it a lot, because to me, it can be a relief actually … If it comes, then I'll go, I'll just accept it. I don't even want to try to fight it, because if it's time, then [I'll] just go."

Ms A, early 20s, SMA
SMA Type 3

Worries and Concerns - Premature Death (Living Purposefully)

"I'm not worried about death lah. But I just hope, I just want that if it happens, it happens peacefully and not … If possible, just take me straight. So, as long as I am here I … I should be … (silence) … concerned about whether I am living my life the way that God wants me to live. Because as long as He doesn't take me away, He has a purpose. So the question then becomes whether I'm living out that purpose."

Mr E, late 20s,
SMA Type 2

"I don't think about it at all."

Ms B, early 20s,
SMA Type 3

"I don't want to live in bed - I don't like that. I … my head is a very complicated place, I cannot keep still. Um … so I have spoken to people about … (pause) … the idea of death or assisted death. It's a very taboo thing in this country. Especially when you have no control over the disease … (silence) … It's a conversation that needs to be had, but every time somebody brings it up, it's always this phrase 'No, you have so much to live for', and I'm like, I don't want to live in bed (laughs), it's boring. I am not worried about death. I am worried about being an invalid."

Ms C, early 30s,
SMA Type 2

"I am not worried about death. I am worried about being an invalid."

3.2.4.9 Worries and Concerns - Difficult Treatment Choices

Not all the FGDs and IDIs covered this topic. However, below are some of the responses on the topic when it was discussed:

"Have discussed with husband, but not certain what to do regarding options for intubation or tracheotomy for ventilatory support. Have not had the opportunity to discuss or share with other families regarding this issue."

Mrs KK, mother of a teenage boy with
SMA Type 2

"I will think like, whatever the treatment is, I want to try it. But then my husband just tells me like, because of the treatment, you want to make the whole family's life miserable? What's the point?"

Mrs. MM, mother of a young boy with
SMA Type 3

3.2.5 Future Hopes and Wants

The hopes and wants for the future expressed by the participants can be grouped under several themes:

3.2.5.1 Future Hopes and Wants - Access to Treatment and Clinical Trials

In terms of new treatment, all the PWSMA welcome anything that will enable them to improve their functional abilities and/or slow down the deterioration of their condition, as shown by the two quotes below:

> "Anything that enables me to gain back the ability to take care of myself or helps to slow down the deterioration of my condition and to manage in terms of taking care of my daily life."

Ms D, early 40s,
SMA Type 2

> "Based on my own knowledge, Spinraza is not a full cure, it's just a drug that helps show improvement. I would like to see something that is a real cure. I think Spinraza is a start - it's a start of something. Yeah, I would like to gain access to all of this, so that I can improve my health.

Mr G, early 20s,
SMA Type 2

However, if the new drug or treatment is provided through clinical trials, some of them have this to say:

> "If such a trial suggests that it could potentially improve my condition, I don't think I would want to participate as it's not meaningful to me. If my current condition can remain like this without further deterioration, I am fine. Unless you tell me that any future clinical trials could be of significant research for future generations. If you ask me to participate in such trial, [then] I would be willing to try on behalf of others."

Ms D, early 40s,
SMA Type 2

> "Yes [to participating in clinical trials], but depending on how bad the side effects are. And hopefully, the medicine can regenerate my cells … slow down the degeneration, improve the function.
>
> "If it's free, yes. If we have to pay for it, then we won't consider it unless maybe we can find a sponsor finance-wise."

Ms A, early 20s,
SMA Type 3

The following was expressed in two separate FGDs with caregivers:

> "Yes, hope, hope for whatever suitable treatment. We now know Spinraza, and besides that, there are a few types of therapy. Gene therapy is out there too. Hopefully, one day, it will be a reality for all SMA patients to obtain the treatment and to improve and better their lives.

Mrs VV, mother of a preteen boy with
SMA Type 3

> "My suggestion and top priority for the Government is to bring in Spinraza. And put aside a certain allocation, you know, every year for SMA patients."

Mr DD, father of a preteen boy with
SMA Type 3

Parents however, also indicated uncertainty when approaching or accepting treatment or corrective surgery, as said by one father:

> "There are factors to consider when making decisions about using treatments, [including] safety, the reputation of the health professional who administers it."

Mr AA, father of a man in his early 20s with
SMA Type 3

This view is echoed by a mother, **Mrs SS** mother of a school-age boy with SMA:

> "My concern is his scoliosis. When he needs to do surgery, which is maybe five years down the road, where is he going to do it? Because in Malaysia, they are not capable of doing it, for patients like [my son] because anaesthesia is a concern. The anaesthetists here are not … confident enough to do surgery for [my son]."

The side effects, length of time and cost involved are also issues of concern as raised by several parents, including **Mrs LL**, mother to a woman in her early 20s with SMA type 2:

> "Whether after taking [a treatment], will her condition worsen or will she have new concerns or complications? And how long is she going to take the medication? Then what is the cost of the treatment, the duration is it going to be whole lifetime kind of treatment? Those are the main things.
>
> "Also, whether the treatment is available in Malaysia or if we have to travel to other country to seek treatment?
>
> "[And] what would happen to her after five years of taking the medication, would she have to face new problems?"

Meanwhile, **Mrs WW**, who lost her first child who had SMA type 1 at six months of age, expressed the following dilemma:

> "If it's for type 1, I hope the medicine or treatment can make the baby live longer. I mean, live longer a bit lah. But it's very selfish [on our part, as] they are suffering a lot, you know. They cannot breathe, they cannot move. You want him to live longer, bur you're like very susahkan (making life difficult for) the baby also sometimes. Very confusing - I also don't know."

Nevertheless, the desire to have done their best is at the heart of the issue for every parent, as what **Mrs YY**, whose child with SMA has passed away, said:

> "Maybe, like [the FGD moderator] has explained, even if we use BiPAP, there will not be much improvement. But I would feel guilty as well. Because if we can, I want to give him BiPAP. Maybe, even if his lifespan is not long, we would have at least helped him."

The question of carrier screening was also brought up to some of the caregivers and the responses were as follows:

> "Oh yes. I will support that and I would have been glad to have been offered that. Also, for those who just got married, if they have not done so - or even before they get married - check your blood as you may be carriers of SMA."

Mr AA, father of a man in his early 20s with SMA Type 3

> "Pre-marriage testing should be incorporated. Pre-marital SMA gene-testing should be like thalassaemia screening now."

Mrs XX, mother of two children with SMA Type 1

"Pre-marriage testing should be incorporated. Pre-marital SMA gene-testing should be like thalassaemia screening now."

3.2.5.2 Future Hopes and Wants - Holistic Post-Diagnosis Care

In addition to treatment, participants expressed their hope to receive more support in terms of medical care, and financial support in terms of equipment needed to enable the functioning of their SMA children.

In one FGD, **Mrs SS**, mother to school-age boy with SMA Type 1, highlighted the following:

"Respiratory care is the most important because that is how it affects the SMA patients. You can have like, a lot of therapy, physio[therapy] and OT [occupational therapy], but the minute you have pneumonia, that's when you need to step up all the care, the protocols. And that's the most important because pneumonia is life-threatening for children, and so, it's a must to take care of the respiratory system first.

"The doctors, they don't know much, and they don't believe that the caregivers or the parents can do more or want to do more. The doctors do not really care enough to want to provide what is needed by the child."

Mr DD, father to a preteen boy with SMA type 3, said:

> "I wish the doctors could be more open-minded in terms of their passion and willingness to understand. I mean they have to understand the parents' feelings and try their best to treat the patient before [giving up] ... Don't say something like, 'there's no hope', 'be ready for this' - I always hear this, you know. I have experience with doctors abroad, which was totally different. So they have to be more positive than negative."

Mrs XX, mother to two children with SMA, also mentioned that there is a need to provide families with technical support:

> "Machines like cough assist are required to help them cough. Others are machines [for] monitoring and nebuliser. So parents need information on how to use the equipment."

Mrs SS adds:

> "Doctors really discourage mothers from helping the child further. When you see the diagnosis is type 1, they write it's difficult, you don't have to do anything more, so they are not giving the BiPAP. In terms of taking care of the child, the BiPAP will monitor the heart rate, the breathing pattern and all that. And if anything occurs, the BiPAP will clearly have all this information, with the oxygen going in. But to the doctors, having the BiPAP is [unnecessarily] prolonging the life of the patient. They don't see that this is what the parents want.
>
> "And work with the parents. Yes, we understand that the doctors cannot know everything, but try to work with the parents. Trust the parents or listen to what the parents want first. Don't see our child as a disorder, see our child as a human first."

Future Hopes and Wants - Holistic Post-Diagnosis Care (Provision of Palliative Care Support)

In an FGD, Mrs SS, mother to a school-age boy with SMA Type 1, made an impassioned plea for the need to have palliative care:

> "I think palliative care is very, very important. The palliative team needs to come in and talk to the parents. Even if I know my child is going to pass on, what is the best I can do during this period? The palliative team needs to assess - these are the things we want to do. If not, even if the doctors keep on resisting and keeping the information away, or discouraging the parents, when the child passes away, the parents will feel guilty and helpless because we didn't do our best. We have cases where parents who are given palliative consultation and all that, feel like, 'Okay, I have done my best, it's okay. That is the journey my child … will finally go on.' If the parents are given that kind of counselling, besides genetic counselling, it gives a very good feeling, as if you are prepared, you have tried your best. Doctors should not discourage parents if they want to do something. They will feel very guilty. There are NGOs now who are willing to support this, so why are the doctors so resistant?"

Future Hopes and Wants - Holistic Post-Diagnosis Care (Professional Mental Health Counselling)

In a similar vein, the need for professional mental health counselling was put forward:

"Counselling for grieving parents. I mean, if the parents can talk to a professional counsellor, a person who is totally impartial and someone professional to listen, you know. I think it's important.

"I have parents calling me who have totally given up on doing anything for their child with a rare disease - not SMA only, but also other rare diseases. They don't even go to the hospital to take the medicine for the child anymore, because in their mind, they think nothing can help their child any more.

"So, when parents have this kind of mindset, who is suffering? The child.

"Maybe the parent is actually going through depression. I think it needs to be addressed. Certain types of counselling for parents with children with special needs, that we can, you know, get referred to."

Mrs SS, mother to a school-age boy with SMA Type 1

Future Hopes and Wants - Holistic Post-Diagnosis Care (Respite care)

As highlighted in the section on the impact of SMA on patients and families/caregivers (3.2.3), the all-consuming nature of caring for a child with SMA has left many parents exhausted and burnt out. #The need for a system of respite care to address this issue was raised in a mothers' FGD by **Mrs SS**, mother to a school-age boy with SMA Type 1:

> "We are very tired! And if the parents or the caregivers are not taken care of, it doesn't only affect the child with SMA, but the whole family.
>
> "We are only hearing of parents who are capable, who can come out and do things themselves; there are many who are not able to do the same. Our hope is to have support for caregivers like us. The Government or other NGOs can come in with a system that actually supports caregivers, like what they do in Australia, where maybe once in a week or once in a month, someone comes in to help relieve the caregivers, which I think is very, very important."

Future Hopes and Wants - Holistic Post-Diagnosis Care (Support Group)

Many parents found that having a support group helped them tremendously and recommend for such groups to be publicised, as said by **Mr GG**, who has an school-age-old son with SMA type 2:

> "This WeCareJourney should be publicised to all the hospitals, so that the doctors know that if there is a new diagnosis of SMA, they can straight away recommend them to join this group and get support. We are supporting each other. From there, we are motivated. We are not alone."

There was a suggestion from **Mrs WW**, whose child passed away from SMA type 1, during an FGD for mothers, to have a support group for caregivers of children with SMA type 1:

> "Have another group that is for SMA type 1 and those who have babies who have passed on. Because type 2 and 3 are very different from type 1 babies - the care is different."

This was emphasised by **Mrs XX** on the role of support groups:

> "There is a need for a support group to share experiences and help teach each other."

Future Hopes and Wants - Holistic Post-Diagnosis Care (Personal Assistants for Adults with SMA)

In one FGD, a mother mentioned the need to have a system of personal assistants to enable adults with SMA to live independently. The experience shared by the adults with SMA in this study who live away from home, indeed showed the importance of having a personal assistant. However, as highlighted by those who engaged a personal assistant, these tend to be foreign workers with expensive contract fees.

> “Perhaps the Government could implement more lenient regulatory terms and conditions to promote recruitment of these skilled foreign domestic workers to assist SMA patients’ needs, and also, reduce the employment contract fee as I need to pay a huge lump sum of money for that purpose. I suggest that the Government organises more helper [personal assistant] training at public hospitals, to let more people be equipped with some basic caregiving knowledge. This training could focus on how to look after patients - not necessarily just SMA patients, but any patient with chronic disease in the household that needs similar caregiving assistance. For example, in the hospital setting itself, there are many staff with different job grades. I really hope to see more of those nurses with white uniforms being employed [in these caregiving roles] as their tasks are mainly to assist patients, like assisting patients to turn their body or assisting the patient to sit up.
>
> “In addition, when I go out from home with my helper, she is supposedly entitled for certain fee rebates. But people have refused to give the half-price discount to my helper and the reason given was that she is a foreign worker and not entitled for any discount. Even though she is a foreign worker, her role is to take care of me, but they are reluctant to grant her any fee rebate.
>
> “When I explained to the personnel, he told me that I have a motorised wheelchair to move around in. I mentioned that at times, my head would drop unintentionally. They could not understand my circumstances; they seemed puzzled and questioned me as to why my head would drop and why couldn’t I use my hand to reposition it. Many of them do not seem to possess any knowledge about the symptoms of SMA.
> “So, am hoping that the Government can provide us with a special permit card that specifies SMA patients are severely disabled persons entitled for special assistance to be given by public staff.”
>
> **Ms D, early 40s,**
> SMA Type 2

“ Perhaps the Government could implement more lenient regulatory terms and conditions to promote recruitment of these skilled foreign domestic workers to assist SMA patients’ needs. ”

3.2.5.3 Future Hopes and Wants - Call on the Government

Future Hopes and Wants - Call on the Government (Increase Awareness)

Several participants called for public awareness across the board and for the public to get to know people with SMA.

> “To them, some of us may look rather normal, but they don’t know that we cannot even lift our hands. When they see us, they think we can do things by ourselves, they think that we are normal, but we really do not have much strength to do that - we need their assistance.”
>
> **Ms D, early 40s,**
> SMA Type 2

> “They should do a census, like if you know that there is a home or a house that has a disabled person, go in and do an interview with them. Talk to them and ask them what they need, what they want. I think that’s important. No one is asking, they only do that in some parts of the rural areas.”
>
> **Ms A, early 20s,**
> SMA Type 3

Future Hopes and Wants - Call on the Government (Improve Accessibility and Inclusivity)

The hope for improving the lives of PWSMA, as well as their caregivers, was passionately voiced in all the IDIs and FGDs:

> “I think the number one thing would be to make places more accessible. Some of us are on wheelchairs, some of us use stretchers, but I think it’s important to make every place accessible, so as to make our life easier. I mean, they have no idea how important it is - a small thing such as a ramp can make such a big difference, you know. Yeah, I think Malaysia is still so bad [in this] - so many places I go to are terrible, like huge, huge steps and all (laughs). I just wonder why is it here like this! I mean that’s kind of independent living, you know. If everywhere was accessible, I could actually call my friends, I wouldn’t have to rely on my parents or my sister.
>
> **Mr G, early 20s,**
> SMA Type 2

> “If you have a system that appreciates you as a human being, you won’t be driven to the corner by people who don’t know what or who you are. There will be no stigma because stigma has been eradicated by simple laws against discrimination. So, law is very important. The practice of law and the enforcement when it comes to human rights. It’s an ongoing process; I don’t think we’ll ever stop.”
>
> **Ms C, early 30s,**
> SMA Type 2

> “I read a quote that said, ‘It’s not the disability that disables us, it’s the society that disables us.” If everyone was given an equal job opportunity, an equal chance on education, I don’t think that will be a problem for me right now. Flexible work hours are actually a must because of our condition.”
>
> **Ms A, early 20s,**
> SMA Type 3

Future Hopes and Wants - Call on the Government (Improve Medical Care Services for SMA)

Several participants raised the idea of a specialised medical institution for rare diseases to help improve medical care for such diseases.

"It is better if it is possible, but depends on the ability of the Government. Like we have the heart disease centre in IJN, we could have a hospital for rare diseases - diseases that are not usually seen."

Mr FF, father of a woman in her early 20s with
SMA Type 3

"Recommend the Government to have a holistic care centre from diagnosis to treatment and support."

Mr BB, father of two deceased children with
SMA Type 1

"I am hoping to see if one day, an SMA care building could materialise. Within it, there could be a specialised centre for SMA patients, comprised of good treatment and other facilities. And similar to a hotel structure, there could be a number of different-sized rooms to host different events like psychoeducation for parents and guardians, and providing skilled training for caregivers, so that all SMA patients could seek proper treatment at one main site.

"It should serve as a learning hub. First of all, when the SMA-affected family registers there, the parents need to receive some form of education regarding SMA and how to look after their SMA kid. Then the parent needs to impart this knowledge of caregiving to their helper on how to manage their SMA kid. It should even have a hostel for SMA patients to live in, and even to start working. With such a centre, it will motivate all SMA individuals who might feel hopeless prior to this; to regain some hope instead of idling [and] just waiting to die like those at their terminal stage. SMA patients are still able to work and accomplish a lot of tasks."

Ms A, early 20s,
SMA Type 3

Concluding remarks

SMA has profound economic, emotional and psychosocial consequences for the patients, caregivers and families. Salient themes relating to diagnosis, care and support of families with SMA children and adults, the impact of living with SMA, and the worries and concerns of both PWSMA and the caregivers, will be synthesised with the findings of the quantitative component of the study in the next chapter.

"I have gone through experiences of injustice and discrimination, so I don't really put up with any stigma that I face outside. I would usually confront the person and I would just tell them upfront that I really need this thing and ask if they can help me with it. Or if anybody stares, I will stare back at them. I don't know why people get used to the fact that they can get away with extreme behaviour when they see a disabled person. I think it's not right, because disabled people are human too. They deserve to be treated with the same respect and dignity like everyone else. I think people need to be educated about disability."

Ms A, early 20s,
SMA Type 3

It is the whole system in Malaysia. It is systemic discrimination!

Ms C, early 30s,
SMA Type 2

Chapter 4
Discussion and recommendations

This final chapter draws together the quantitative and qualitative findings of the study, highlighting salient issues, themes and the implications for health and social care professionals, as well as policymakers. A number of key recommendations will also be presented.

4.1

The Journey to A Diagnosis

From our study, which is consistent with the results of previous research (Qian 2015, Lin 2015), the journey to receiving a diagnosis from the time symptoms were first noticed, was often protracted and fraught with frustration and anxiety. Not uncommonly, caregivers consulted with many different healthcare professionals in their search for a diagnosis, who often took a "wait and see" approach or gave false reassurance. This approach was usually compounded by poor awareness and recognition of SMA symptoms, a poor sense of urgency in facilitating referrals to the appropriate specialists and not taking serious consideration of the problem, while also casting aside and dismissing the caregivers' concerns.

At the same time, caregivers had to independently seek more information about SMA as it was not readily offered to them, and access to trained, compassionate care and services was often come across via word of mouth or by chance. Many participants abhorred how information was sometimes conveyed in a crude, uncaring and insensitive manner, which further frustrated and distressed the parents. The negative experiences related by the participants is a consequence of the challenges faced by doctors and the limitations of the healthcare system in Malaysia. Rare diseases are seldom covered in depth during medical school as they are indeed rare, resulting in lack of adequate exposure to and education on diseases such as SMA. Systematic limitations imposed on healthcare providers who already have a heavy workload with high number of patients and lack of resources or specialists to refer to, contribute to the negative experiences undergone by the participants, especially so if the SMA patients were diagnosed in the early 1990s. Caregivers to younger SMA patients related less arduous and less traumatic experiences compared to older SMA patients and their caregivers.

After being given the diagnosis, families struggled to balance leading a "normal life", while coping with feelings of grief and continually seeking support and ways to relieve the PWSMA's symptoms, while also dealing with various mental, emotional, financial and social issues, including the spectre of an uncertain future and premature death.

"The negative experiences related by the participants is a consequence of the challenges faced by doctors and the limitations of the healthcare system in Malaysia."

4.2

The Impact of SMA Symptoms

The qualitative findings highlighted and emphasised in greater depth and breadth what was reported in the quantitative survey on the symptoms that most impacted PWSMA's and their caregivers' lives. In this study, the foremost issue was the lack of independence and mobility. In particular, this was a critical milestone for patients with SMA type 3 as they transitioned from being independently mobile to becoming wheelchair-bound. On the other hand, wheelchairs are a part of life for those with SMA type 2, who are usually adept at utilising powered or electric wheelchairs. However, all face difficulty travelling independently or navigating their environment due to sub-optimal infrastructure and public amenities that are inaccessible to wheelchairs.

Other examples of activities of daily living that were impacted are transferring oneself (to and from wheelchair/scooter to bed or toilet), attending to personal hygiene independently and turning in bed. Muscle weakness, joint contractures, fatigue and lung infections were symptoms that caused major impact to their quality of life. The progressive muscle weakness of the disease eventually leads to losing upper limb mobility and to the development of upper and lower extremity contractures, largely due to limited mobility. Simple tasks or activities like moving an arm, lifting up one's head and using utensils to eat will require ever greater exertion over time, making fatigue a constant part of their lives.

"Muscle weakness, joint contractures, fatigue and lung infections were symptoms that caused major impact to their quality of life."

One overriding concern for caregivers of SMA type 1 patients is the life-threatening respiratory-related symptoms (e.g. difficulty breathing, inability to cough and clear secretions, and lung infections). The all-consuming nature of having to maintain constant vigilance to ensure the airway passages are always clear, and the overwhelming anxiety - to the point of obsession - over risks imposed by infections, significantly impacts their lives and interpersonal relationships. Furthermore, adults with SMA type 2 reported suffering from difficulty expectorating phlegm with lung congestion. This is a perpetual struggle and preoccupation throughout their waking hours, as well as while sleeping. This in turn results in considerable disruption of their daily activities, work and sleep. In this regard, a cough assist machine can markedly ease a patient's breathing effort through improving one's coughing reflex and assist in more effective clearance of secretions. As shared in the qualitative part of the study, an adult respondent with SMA type 2 elaborated on how the cough assist machine transformed his life by improving his ability to focus on his work and graduate studies.

However, necessary equipment to improve the quality of life of PWSMA, such as the cough assist machine, mobility aids like wheelchairs, and home renovations to accommodate wheelchairs, are not covered by insurance and frequently result in tremendous financial strain on the families of the PWSMA. This is further compounded by frequent work disruptions to tend to the PWSMA, as well as lack of appropriate, disabled-friendly employment opportunities for adult PWSMA. All this, in addition to the limited social activities available to PWSMA due to access and financial issues, contributes to tremendous physical, emotional, psychosocial and financial burdens on both PWSMA and caregivers, as communicated by many participants in this study.

4.3

Psychosocial Well-Being

As shown in Chapter 3, 76% of caregivers and 38% of PWSMA reported experiencing anxiety. Slightly over half of caregivers and PWSMA reported going through stressful experiences, while 20% of caregivers and 38% of PWSMA reported feeling depressed. While DASS 21 scoring did not yield findings, which warrants particular concerns on the participants' psychological well-being, the qualitative data provided much insight into the psychosocial adjustment of the PWSMA and caregivers.

The long-term, progressive nature of rare diseases like SMA requires a lifelong family-centered framework of caregiving. Parents may have to function as long-term primary caregivers even after their child enters adulthood. This is especially as there are many personal, societal, medical and institutional challenges that impede the transition to independence for young adults with SMA. Individuals and families with SMA face numerous multi-faceted challenges associated with the complexities of care needs, assistance in everyday responsibilities and upkeeping of psychosocial health for all concerned, and families must work to incorporate these challenges and solutions into their family life for years to come.

While all the participants demonstrated great strength and resilience in their ability to cope with the stress and strain of living with SMA or caring for one with SMA, we were able to discern a number of stress points and pitfalls using a lifespan perspective, as the PWSMA and caregivers navigate their lifelong journey of dealing with SMA.

76% of caregivers and **38%** of PWSMA experienced anxiety

4.3.1 For PWSMA

For PWSMA, growing up feeling or being told that they are different - with the implication of being inferior or abnormal -has had an impact on their self-esteem. Growing up being loved or rejected in a nuclear or extended family environment has a far-reaching impact on the PWSMA's sense of well-being, as illustrated in the personal accounts shared in Chapter 4.

Entering school is another critical event in the life of PWSMA. The experience of being isolated, discriminated against, or even bullied, is a reality for some of these individuals, and is undoubtedly a stressful, and potentially traumatic, experience for any child. Conversely, being supported by peers and/or teachers can go a long way in the person's ability to form positive interpersonal relationships in life.

The findings of this study corroborated with previous research on the importance of family care, extended family support and friendship in the development of adolescents and young adults with disabilities (UNICEF 2017). Having some degree of income security and a comparable social life to those without disability were other areas that appeared to make a difference in the psychosocial well-being of PWSMA.

“ The experience of being isolated, discriminated against, or even bullied, is a reality for some of these individuals, and is undoubtedly a stressful, and potentially traumatic, experience for any child. ”

4.3.2 For Caregivers

It is noteworthy that a higher percentage of caregivers reported experiencing anxiety (76%), compared to PWSMA (38%). Indeed, qualitative data revealed that anxiety is very much a part of daily life for caregivers. Anxiety permeated their experience, starting from when they noticed “something not right” about their child up to receiving the diagnosis, and continued throughout the daily care of the child, as well as spilling over into their relationships with family and friends.

The uncertain course of SMA is exacerbated by foreknowledge for some families who have more than one child with SMA and have already faced the death of one. The anticipatory grief of having to face the death of another affected child and going through another cycle of desperation in caring for the remaining affected child(ren) contributes to feeling overwhelmed and anxious. However, only a single caregiver has sought professional mental health counselling and none have received palliative care referral or support, despite SMA being a life-limiting illness. A discussion on palliative care early in the course of the disease is appropriate, but these discussions may not occur spontaneously as parents in general do not initiate such discussions with their clinicians, even though they desire to have them. This finding emphasises the importance of healthcare providers' initiating discussions on palliative care.

Qualitative findings also highlighted the strength and resilience of many parents in coping and supporting their children - in some cases, adults - with SMA. Nevertheless, as expressed in nearly all the FGDs and IDIs, the need for psychosocial support at different stages of their journey is highly recommended and will be elaborated on later.

“ Anxiety permeated their experience…. ”

4.4

Education

Issues related to education featured prominently under the section on Impact on Patients and Families/Caregivers (3.2.3) in the qualitative data. Educational attainment for PWSMA was varied, with one never having attended a formal school and another who had to quit after primary school because of deteriorating strength, to four who have completed or are completing tertiary education, as well as one pursuing a graduate degree. This clearly shows that given the opportunity, PWSMA can excel in academic pursuits, just like any other person.

The schooling experiences of PWSMA bring to the fore many unresolved issues surrounding education for children/persons with disabilities, and specifically for PWSMA, in Malaysia. Caregivers lamented their negative experiences of having to search for or apply to schools that would be able to cater for the special needs of their children, as well as being denied admission to schools, including private ones. It is disconcerting to note that this situation does not appear to have changed significantly over the years, as similar issues were experienced by PWSMA and their caregivers more than a decade ago. Clearly, there has not been much inroad in applying the principle of "educational for all", despite the signing of the Salamanca Statement (UNESCO 1994), the principle of inclusivity in the Education Act 1996 (Government of Malaysia, 1996) and the passing of the Persons with Disability Act (2008), which calls for accessibility to all aspects of life, including education, for persons with disabilities (PWD). Furthermore, Malaysia's National Education Blueprint (2013-2025) has set a target of 75% of special needs students receiving basic education via the Inclusive Education Programme (IEP) by 2023 and the more recent Zero Reject Policy, which needs to be translated into action at the ground level.

> "Schools were reluctant to admit, or even outright rejected, children with feeding tubes or who needed a ventilator in between classes"

An issue that emerged clearly from the qualitative findings was that children with SMA were often thought of by school authorities as requiring special education (for those with learning disabilities), despite most of them being of normal intelligence and not having any learning disabilities. These children were then either refused admission to the school altogether or placed in special education classes. Schools were reluctant to admit, or even outright rejected, children with feeding tubes or who needed a ventilator in between classes. This is despite the parents offering to be present at school to help support their child's needs. In some cases, teachers were not accommodating to the PWSMA's needs and indicated that having to attend to these needs when necessary would disrupt the class. A number of participants who did gain entry into schools experienced being first placed in special education classes, then only allowed to join regular classes after a certain years.

Some also shared experiences of feeling unsupported, being discriminated against, and even bullied, although a few others were happy with the "special" attention and support they received from school authorities. Some parents reported feeling distressed upon seeing their child being left alone or isolated from fellow students during recess, classes on upper floors or pick-up time. However, some participants had positive experiences of being helped by fellow classmates who carried their books, bought food from the canteen for them during recess and pushed, or even carried, them in their wheelchairs.

4.5

Employment

Workplace discrimination is an ongoing issue in Malaysia and is particularly rampant against marginalised populations, especially PWD. PWD often face discrimination during the recruitment process and are not provided with equal job opportunities and considerations, thus negatively affecting their lives and ability to be independent.

Adult PWSMA bemoaned the lack of disabled-friendly work environments, i.e. wheelchair-accessible and equipped with disabled-friendly toilets. This is despite tax incentives to private companies that train and hire PWDs, as well as modify their facilities to accommodate the needs of PWDs. In addition, the government introduced a policy providing for a quota of 1% employment of PWDs in the public sector in 2010. However, there is still a lack of accessible infrastructure in most workplaces and employers remain unwilling to allow the flexible working hours required for PWDs to contribute effectively.

> "PWDs often face discrimination during the recruitment process and are not provided with equal job opportunities and considerations"

4.6

Medical management & treatment options

Many families and caregivers of PWSMA were lost navigating the complex and fragmented array of services associated with the transition of care from childhood to adulthood. A well-coordinated and multidisciplinary team (MDT) approach is crucial for optimal evidenced-based standards of care for a life-limiting neurodegenerative condition like SMA. Half the PWSMA attend neurology and respiratory clinics. At home, cough assist and suction machines are commonly used to remove secretions, and those requiring respiratory support use non-invasive ventilation. A care pathway with anticipatory guidance throughout the patient's lifetime would be useful in monitoring PWSMA in order to prevent and diagnose early complications and initiate prompt treatment.

> “ One element unique to caring for children and adolescents with rare diseases like SMA is the need for appropriate transitions of care ”

Scoliosis in SMA is directly linked to the severity of the type of SMA. The lifetime probability of scoliosis surgery is high in SMA types 1c and 2 (approximately 80%) and depends on the age of loss of ambulation in type 3 (Camiel 2019). There are two major consequences that follow the worsening of scoliosis with the deterioration in muscle strength. Firstly, development of restrictive lung disease compounding the breathing difficulties caused by weak intercostal muscles, and secondly, stiffening of the joints and consequent painful joint dislocation or subluxation, caused by reduced mobility. Progressive scoliosis can impact the ability of patients to sit comfortably, be cosmetically unappealing, and in severe cases, exacerbate pulmonary disease. Non-operative treatments have not been shown to be effective at preventing the progression of scoliosis. The main goal of operative treatment is to improve sitting balance and prevent progression of scoliosis (Garg 2016).

However, only three PWSMA reported regularly attending orthopaedic clinics, and four out of 13 have undergone scoliosis surgery. The lack of local expertise or lack of confidence in such expertise, and the perceived risk of such surgery, deterred other caregivers and adult PWSMA from undergoing the operation. The majority of studies has found that spinal surgery can maintain and improve breathing function (Chou 2016), reduce the frequency of chest infections, improve tolerance of sitting still for long periods (i.e. in the wheelchair and in school), and improve self-confidence and quality of life. These findings entail further exploration and education amongst caregivers and PWSMA on the benefits of surveillance and timely intervention for scoliosis.

Three PWSMA attended allied health specialty clinics, with the most common interventions or therapies being physiotherapy, occupational therapy and hydrotherapy. A participant deplored the state of the physiotherapy department in a government hospital, which constantly had long queues. This is hardly avoidable as a government hospital offers medical care and services at almost next to nothing, which inevitably overwhelms the system with too many patients. Additionally, the general experience of the participants was that the therapists were not knowledgeable about SMA patients, and thus unable to provide individualised rehabilitation programmes, instead, prescribing exercises for stroke patients. Feelings of demotivation eventually lead to patients dropping out of these therapeutic sessions.

One element unique to caring for children and adolescents with rare diseases like SMA is the need for appropriate transitions of care, be it with regards to specific needs, within the home, in school or to adult clinical services. Transitions along the disease trajectory create many challenges and must then be integrated into supporting the child in living as “normal” a life as possible. Examples of these transitions include the need to start using a wheelchair, feeding tube or mechanical ventilation. Transitions may also occur within families, such as when a caregiver can no longer provide the necessary care, requiring someone else to take over the role.

This is hardly avoidable as a government hospital offers medical care and services at almost next to nothing, which inevitably overwhelms the system with too many patients.

4.7

Finance and Insurance

Substantial healthcare costs are associated with the management of SMA. Such costs come in terms of hospitalisations, emergency consultations, frequent visits to various subspecialists and out-of-pocket costs associated with medical care and equipment (Julio 2017). The expense of purchasing assistive devices, mobility aids, respiratory support equipment (e.g. ventilators, BiPAP, CPAP and cough assist machine) and home modifications are exorbitant and are not covered by insurance, forcing some parents to find a second job, take out loans or look for charitable assistance. Mothers usually have to leave their job to become a full-time caregiver, leading to a further loss of income and added financial responsibility and stress on the other parent.

Additionally, due to lack of employment opportunities, most adult PWSMA are unable to be financially independent. Even if they are able to find employment, the available jobs often do not pay well and their income is often consumed by the need to hire a full-time maid or personal assistant. Frequently, PWSMA need to continue to rely on family and social services for support.

Financial woes were an overarching concern for all PWSMA and their families.

> “ Substantial healthcare costs …..not covered by insurance..”

4.8

Recommendations

4.8.1 Strengthen Clinical Management for Rare Diseases

While SMA may be considered a rarity by most clinicians, new advances in SMA management, such as the emergence of novel therapies, has escalated the need for SMA education and awareness among all levels of healthcare providers, especially paediatricians and general practitioners. Untreated SMA can have devastating consequences. Timely identification of symptoms and prompt referral to a specialised centre is critical to avoid a long diagnostic odyssey and enable earlier access to evidence-based care and intervention, as well as informing reproductive planning and reducing patient and parental stress.

4.8.1.1 A Systematic Approach to Diagnosing SMA

Rather than relying on the opportunistic identification of patients or carriers in a clinical setting, the reported delays in diagnosis of inherited conditions that present with developmental delay support the consideration of a systematic approach to diagnosing SMA at a population level.

Prenatal diagnosis (PND)

Slightly more than half of the caregivers have heard of PND and 40% would consider this service in the future. Ten percent have undergone invasive PND and all of them were very satisfied with the service.

There is an alternative to this traditional invasive prenatal testing, which is a fast, low-cost, non-invasive maternal blood test for prenatal screening of SMA for families with a history of SMA. This test allows identification of affected foetuses within the first trimester.

Undeniably, there are many ethical and moral dilemmas regarding the termination of pregnancies (TOP) with severe foetal conditions. While TOP can be a medically-guided decision based on the judged danger to mother and/or foetus, it is also a personal choice of the mother that is heavily influenced by socio-cultural values, religious beliefs and legal rules.

Carrier screening

Population-based SMA carrier screening is to identify couples at risk of having a child with SMA, thus allowing informed reproductive choices. Carrier screening is ideally offered to couples when planning for a pregnancy, as this would provide reproductive options to them.

Increasingly, carrier screening for panels of genetic conditions, including SMA, are being offered. Population-wide SMA screening to quantify the SMN1 copy number is recommended by the American College of Medical Genetics and Genomics (Xiao 2020).

Preimplantation genetic diagnosis (PGD) offers a realistic option for having an unaffected child without having to resort to PND and the dilemma of therapeutic abortions. Our study revealed that only 8% of PWSMA and 28% of caregivers have heard of PGD, but would consider such a service in the future. Further steps are needed to increase awareness of PGD and ensure that this option is shared with couples seeking genetic counselling.

Newborn screening (NBS)

NBS has been widely used over many decades to identify children with various life-threatening conditions. In infants with SMA type 1, the onset of irreversible denervation occurs within the first three months of life, with 90% of motor nerve units lost by six months of age.

Specific novel treatments, including gene therapy for SMA, are already available, but at a high cost that is not affordable by many healthcare systems. However, NBS may allow an affected child to be enrolled in a clinical trial or highly competitive global pre-approval access or compassionate use programme before irreversible neuronal loss occurs, thus enabling patients to obtain more proactive treatments. Massively parallel sequencing or next-generation sequencing, accurate carrier screening tools and comprehensive tests in neonatal care can be offered (Feng 2017).

4.8.1.2 Multidisciplinary Team (MDT) Engagement

After a diagnosis of SMA is made, the patient and family face a complex array of medical and psychosocial needs, which should not be dealt with in isolation or silos. An ideal solution would be the formation of a multidisciplinary neuromuscular centre that provides holistic whole-of-lifetime services from diagnosis to treatment, rehabilitation and psychosocial support. Transdisciplinary collaboration and open communication between all the stakeholders of various specialties focusing on patient and family-centered discussions are paramount. It is essential to have a coordinating doctor who is aware of the disease course and potential issues. This role usually falls on the paediatric neurologist or geneticist, who, together with the families, will monitor various aspects of the disease to provide individualised anticipatory care plan and acute care protocols (Eugenio 2018).

Discussion of the patient's and/or family's goals and alignment of goals within the MDT is essential. Balancing caring for the child at home for as long as possible, long-term survival, quality of life, comfort and the availability or lack of resources are important issues to address.

It is also recommended that the creation of a post for SMA family care officer be looked into by the Government. This family care officer should be trained in bereavement counselling and psychological support. At the same time, they can serve as advocates for SMA families and help them navigate the complicated treatment options, tackle school environments and deal with the various challenges that face them with regards to the disease.

4.8.1.3 Respiratory Care

Lung disease is of primary concern as it is a main cause of death in SMA. The type of SMA will determine the extent of respiratory involvement. A proactive approach of introducing respiratory therapies early in the disease is the way forward in treating the pulmonary manifestations of SMA. Hence, input from a good team of pulmonologists and respiratory therapists should be initiated early, focusing on detailed clinical assessments such as cough effectiveness, sleep studies and pulmonary function tests to provide the best level of care for PWSMA. This dedicated team should be made accessible at every level of healthcare, starting from primary care all the way up to tertiary care centres.

4.8.1.4 Orthopedics and Rehabilitation Care

Rehabilitation goals differ according to the different types of SMA and the course of the disease, but these goals should also depend strongly on the patient's and family's wishes. Functional assessment using validated scales and assessment tools is critical before a rehabilitation programme is embarked on. Musculoskeletal and respiratory rehabilitation and orthotic care go hand in hand.

Some of these goals include optimisation of function, minimisation of impairment and prevention of contractures and scoliosis. Nutritional management and emphasis on bone health should also be included. Speech and language therapists and nutritionists have crucial functions in addressing feeding and swallowing problems due to bulbar dysfunction. Additionally, other allied health professionals like physiotherapists and occupational therapists play indispensable roles in the care of SMA patients. Medical advances such as non-invasive mechanical ventilation, cough assist machines and increasing ICU survival have extended the lives of SMA patients. This focus on complex, life-sustaining treatments can delay palliative care. Such treatments prolong life and may add to quality of life as measured by the patient, but they also make it more difficult to predict end of life. However, palliative care is essential for the management of SMA patients, and thus, discussions on advanced care plans and prompt referral to a palliative care team should be conducted soon after diagnosis while the management plan is being formed.

Surviving adult PWSMA struggle with activities of daily living and deteriorating functional abilities contributed by worsening scoliosis and joint contractures and/or excessive weight gain. Establishment of an NGO or community-supported endeavour for the independent living of these individuals, which comes with the provision of trained personal assistants or aides, would help. Basic caregiving workshops can be organised to equip helpers/aides/personal assistants with the relevant knowledge and skills, focusing on how to care for patients with disabilities in general and SMA specifically.

4.8.1.5 Opportunities for Clinical Trials and Novel Therapies

With the advent of new therapies and many recent clinical trials, the survival and natural history of SMA will likely change to an increased survival rate and improved overall function. A national SMA registry is needed to provide better insight into the disease's epidemiology. The availability of prevalence data will help characterise the burden of SMA, ensure patients have access to optimal care, provide research opportunities and ease participation in clinical trials for new and innovative therapies.

4.8.2 Improve Psychological Support Services

There is need for a support system at each stage of SMA as many parents have painfully shared the mental torment, anger, stress, depression and anxiety they go through at each stage of the disease. This includes dealing with the shock and grief that comes with the diagnosis and prognosis, the mental burdens of caring for their child and the grief when the child passes away.

In the first instance, clinicians, counsellors, psychotherapists and social workers need to be equipped with the relevant knowledge and communication skills, including training on how to break bad news and empathy. The process of coping with the disease on the part of families and patients may mean seeking a reason for why this has happened to them. Responses may differ by culture, socioeconomic status and educational level, but having a professional to facilitate the process would greatly help. Professional counselling for caregivers to help address mental health issues is needed. Besides counselling, workshops teaching mindfulness and self-compassionate care (Kristin 2020) can be utilised to help caregivers cope mentally and emotionally. There is also a dire need for a respite care system to help relieve parents of their duties temporarily to allow them some rest.

Having a child or children with SMA presents many additional challenges for parents as a couple and for intra-family relationships. The day-to-day care of a child with a disability tends to fall disproportionately on the shoulders of mothers, which may lead to feelings of being unsupported and isolated. On the other hand, other children in the family may feel neglected. The differences between male and female ways of handling and expressing emotions such as non-acceptance, anger, guilt and fear, can also potentially lead to a rift in the marriage. Professional family counselling to help couples and families address issues and differences arising from the demands of caring for a child with SMA is recommended.

The hereditary factor of genetic diseases like SMA can pose unique issues for patients and families. This includes parental guilt regarding transmission, the potential for prenatal diagnosis, ethical and moral dilemmas with regards to the ability to terminate an affected pregnancy, and foreknowledge of the disease and its prognosis. The difficulty of balancing hope with reality is particularly acute with genetic diseases in light of the rapidly-expanding understanding of genetics that can influence treatment options, caregiving and end-of-life decisions.

A more seamless approach to palliative care and bereavement counselling is also called for. Palliative care support does not mean giving up on life, therefore this approach should be one that avoids unnecessary conflict between palliative care and life-prolonging treatments. The palliative care team can help families with more than one SMA child to resolve grief from the first death and anticipate the grief process prior to the death of the next child. It is also important that bereavement counselling not be limited by time as a number of participants were still grieving for their children who passed away six or more years ago. In addition, spiritual care is of paramount importance and should be provided to help families deal with the uncertainties of the disease and to find ways to accept death as a part of life. The availability of palliative care services and trained personnel is scarce in Malaysia, thus such services need to be beefed up and more personnel trained in this area of care.

Parents feel a moral imperative to be the best possible guardians, yet they often feel that the healthcare system deprives them of that role when an infant or child is very sick. Smith stresses the need for clinicians to engage in meaningful advance care planning discussions, stating, “If we want to honour our patients’ wishes about the type of care they want to receive, we have to first discern those wishes.” (Smith 2015) Through advanced care plan discussions, family may achieve greater control over issues such as where the child dies, how much care the child receives, who will provide care, and very importantly, what type of environment will be created for the remainder of the child’s life.

4.8.3 Empower Patient-Parent Advocacy Organisations

Caregivers reported feeling a sense of solidarity with and receiving moral support from support groups. Support groups can provide mutual aid and information-sharing, which enables parents to feel less helpless and more adequate in caring for their children. Parents of children with genetic disorders may experience ostracism from their own families, who do not want to acknowledge that this disease could have happened or might happen to them. And families, who often feel abandoned after their child with SMA dies, continue to receive an “ongoing sense of security and support” from the group.

Empowering families to be heard and engage in decision-making

It is essential for patients and their caregivers to articulate their needs with regards to accessibility laws, i.e. rules and regulations to facilitate disabled access to transport, infrastructure and public amenities, educational needs such as teacher/student aides, transitioning to adult care and recreational activities within the community.

Collaboration with the education system

Parents, particularly those of children with special needs, should be viewed as vital partners in the education system. They can share crucial information on “what works” for their child with teachers, as well as carry on any teaching needed at home. As parents, their input are necessary in designing their child’s IEP. Close cooperation, mutual respect and teachers who are dedicated advocates of equality in education are key to ensuring the successful implementation of the IEP.

Collaboration with the healthcare system

Patients and families can play an active role in helping to shape national healthcare policies and practices that relate to them. They can help enhance the visibility of rare diseases like SMA at the national and international level to increase public awareness, as well as within the healthcare system as a human rights issue and a public health priority. Previous studies have shown the importance of the continuing role of patient communities in driving the establishment and adoption of legislation and programmes to improve rare disease care.

4.8.4 Educational and School Reforms

The principle of inclusive education

Implementation of the IEP initiative has shown that the Ministry of Education (MOE) is committed towards fulfilling the rights of students with special educational needs to have equal educational access. There has been argument by some quarters that MOE has not fully implemented IEP. Inclusive education needs to be promoted by scaling-up the initiative at all levels (Amar Singh HSS et.al. 2018).

Educational opportunities for all

"Zero reject" is an educational philosophy that says no child can be denied an education because they are "uneducable". The right to education must be translated into action and inculcated into the mindset of educational personnel at all levels. The Zero Reject Policy needs to be implemented with the appropriate accompanying facilities and physical environment.

Strengthening teacher training programmes

All teaching staff must be trained in disability awareness and the basic fundamentals of special education needs. Knowledge on SMA needs to be incorporated into such programmes as a child's progressive disease can have an impact on the school community, fellow students and teachers.

Teachers should receive continuing education courses to equip themselves with the knowledge and skills to teach differently-able children without discrimination and being a good role model in empathy and compassion to their students. Nurturing such attributes among students is an important part of the character-building process, and teachers are an essential component in creating a truly caring and integrated society in which there is a natural co-existence of friendship and classmates rendering assistance without condescension. The MOE will need to scale up provision of early intervention services, increase inclusion programmes within mainstream school settings, and set and improve the standard of support services for special education needs children.

Training in counselling and collaboration

Teachers should also receive mentoring on the psychosocial needs of students with SMA and how to counsel them, as well as their parents. Teachers should be equipped via training with the necessary interpersonal and consultation skills for collaborating with families as this can help make inclusion successful in schools.

4.8.5 Equal employment Opportunities

Anti-discrimination legislations and employment rights for PWDs have moved to the forefront in developed countries. Malaysia has several employment laws in place to address these issues, such as the Employment Act 1955 (Act 265) and Industrial Relations Act 1967 (Act 177). But there is no distinct Malaysian law that fights against the discrimination faced by PWDs. There needs to be a paradigm shift from a charity-based approach to a human rights one in addressing the awareness and recognition of the roles, contributions and future of PWDs in society. Guided by more egalitarian and altruistic principles, the universal human right approach advocates that quality of life and equal opportunity should include PWDs too; and not merely remain the exclusive privilege of able-bodied people (ILO 2006).

Legislate a workplace discrimination law for PWDs

Malaysia has an international obligation to protect job seekers from discrimination, having ratified the UN Convention on the Rights of Persons with Disabilities (CRPD) and enacted the Persons with Disability Act (2008). Perhaps the Ministry of Human Resources can work with other stakeholders and explore how to redirect recruitment and staffing in a non-discriminatory manner.

Proposed amendments to the Employment Act 1995

These amendments aimed at protecting job seekers from discrimination were originally supposed to be tabled in the Dewan Rakyat in October 2019. The way PWDs are marginalised, excluded and deprived from employment further aggravates and compounds their dependency on society - a situation that is neither inclusive nor sustainable in the long run. There needs to be louder voices backed by evidence to inform policy towards forming a socially-inclusive and compassionate society that reinstates and reintegrates the roles of PWDs in the mainstream Malaysian economy.

4.8.6 Redirect Policies and Insurance

4.8.6.1 Clear and Committed Policy Directions

This commitment must first be clearly and firmly espoused by policymakers themselves, who must promote this policy at a national level to all levels of government/civil service, academia and the private sector. We need to leverage the provisions enshrined in the 2030 Agenda on Sustainable Development and the CRPD to ensure that all persons living with rare diseases are not left behind (UN 2019).

Fundamentally, the government needs to endorse national plans for rare diseases that provide frameworks incorporating a coordinated "whole-of-government" approach to rare diseases, as opposed to a piecemeal approach. These plans should outline a cohesive clinical, public health and disability service approach to rare diseases that addresses prevention, timely diagnosis, early intervention, and appropriate access to treatments and rehabilitation.

Such strategies do exist regionally and internationally, and can provide guidance on how current local services can be better integrated with the translation of new knowledge. Recognition and unwavering support need to be given to local experts and their international network. In addition, it is essential to have specific measures such as priorities, actions, budgets and timelines drawn up and followed through.

4.8.6.2 Regulations and Incentives

The Ministry of Home Affairs should consider implementing specific regulatory terms for the recruitment of skilled foreign domestic workers who will work as an aide/maid or personal assistant, providing special assistance to PWDs, e.g. adult PWSMA. These terms can include fee rebates, discounted employment contract fees and special permit cards, among others. The Government should also consider removing or "zero-ing" the tax on imported medical equipment such as powered wheelchairs, cough assist machines and BiPAP, which are essential for the management of SMA, in order to help reduce the financial burden on PWSMA and their families.

Special financial assistance or awards (e.g. a "Caring School" award) can be given to schools that champion and practise inclusion, in order to give due recognition to the school's efforts to embrace, respect and value differences among students of all abilities. The financial assistance is necessary to purchase teaching resources, renovations for better access (e.g. ramps and wheelchair-accessible toilets), and employment of teacher aides and allied health services professionals.

To facilitate social integration and the recruitment of qualified PWDs into mainstream employment, the Government offers employment tax incentives to encourage hiring PWDs. Employers who train and hire PWDS can claim double tax relief for the associated expenses. Additionally, employers are entitled to a tax rebate on the costs of modifying facilities for PWDs. However, awareness is lacking among much of the private sector of these incentives. In addition, the Government implemented a 1% quota for hiring PWDs in the public sector in order to ensure inclusivity beginning in 2010. However, the policy has yet to gain traction as the percentage of PWDs employed by the public sector thus far was 0.23% in 2018.

4.8.6.3 Genetic Non-Discrimination and Insurance

Financial strain is a major concern for families with SMA children as the expenses incurred by hospitalisations and purchase of expensive equipment and treatment are not covered by insurance, but instead, paid out of pocket. This is because, unlike the United States and most European countries, which outlaw genetic discrimination by insurance companies, the Malaysian government has yet to reach a position on such laws. In the US, the Genetic Information Non-Discrimination Act (GINA) of 2008 prohibits insurers from using genetic information to their advantage and deny coverage to a healthy individual or to charge him a higher insurance premium on the basis of predisposition evidence alone, i.e. family history. In an effort to protect our right to affordable insurance, we need to enact laws safeguarding Malaysian citizens against abuse of genetic information by insurers (and employers). The National Bioethics Council, which is entrusted with the task of mainstreaming bioethics and disseminating information on bioethical issues, should initiate open discussion on this topic with Bank Negara Malaysia, which regulates the insurance industry.

4.8.7 Increasing Community Awareness and Education

The way forward to changing the perception towards PWDs, raising public awareness of SMA, and ensuring accessibility and inclusion at all levels of society without discrimination, is through education. This requires dedication and cooperation from multiple stakeholders and various ministries to inculcate in our society feelings of altruism and compassion. Our education system should focus on building an inclusive culture in all schools, workplaces and the community in general, where diversity is embraced, respected and valued. There is a need to develop, gather and disseminate information on SMA and other rare diseases in linguistically and culturally appropriate formats. Patient advocacy groups play an immensely important role in continually promoting these endeavours through collaboration and networking.

4.8.8 Future Research

The majority of the participants were from urban areas and were fairly well-educated. Future studies should aim to uncover the experiences of PWSMA and their caregivers in rural areas and gain deeper insight into sociological areas such as differences in religious and cultural attitudes towards medical interventions and reproductive options. Other areas of research interest include the perspectives and experiences of the healthy siblings of PWSMA, the challenges of adolescents living with SMA and the experience of families who have participated in clinical trials.

Research targeting the general public and healthcare providers to determine the level of awareness regarding SMA and the factors associated with it may be beneficial to help uncover ways to increase awareness of the disease in these groups. The barriers faced by healthcare providers in providing optimal care for their patients can also be looked into to address the challenges within the healthcare system.

Overall, sustained efforts from all relevant stakeholders such as the Government, healthcare workers, policymakers, NGOs, PWSMA and their caregivers are required to bring about systemic change and enable PWSMA to contribute to society, as well as to lessen the burden of living with SMA in Malaysia.

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Appendices

Appendix 1

The impact of living with spinal muscular atrophy in malaysia from patients and caregivers perspective (mysma)

Section A: Demographic information

Please tick ☑ where appropriate.

1. Role:

☐ A person living with SMA Type ☐ I ☐ II ☐ III ☐ IV

☐ Caregiver to a person with SMA Type ☐ I ☐ II ☐ III ☐ IV

Relationship to the person: _____

2. Gender:

☐ Person living with SMA ☐ Male ☐ Female

☐ Caregiver ☐ Male ☐ Female

3. Date of Birth:

_____ (The person living with SMA)

_____ (Caregiver)

4. Year of diagnosis:

_____

5. Marital status:

☐ Single ☐ Married ☐ Divorced ☐ Widowed

6. Highest qualification

☐ No formal education ☐ Primary ☐ Secondary ☐ Tertiary

7. Occupation:

☐ Private sector ☐ Public sector

☐ Unemployed ☐ Pensioner ☐ Homemaker

8. Total household income per month:

RM: _____

Section B: Impact of SMA on daily lives

Please tick ☑ where appropriate.

9. On a day to day basis, primary caregiving for me/my loved one is provided by (max. 2):

☐ Father ☐ Mother

☐ Partner/Spouse ☐ Grandparent(s)

☐ Sibling/other family member ☐ Nurse or other professional caregive

☐ Maid or nanny ☐ Other (friend, other relative)

10. Were you/your loved one ever able to sit independently?

☐ Yes ☐ No

11. Were you/your loved one ever able to walk independently?

☐ Yes ☐ No

12. Which of the following have you experienced as a result of coping with your/your loved ones SMA? Select those are applicable to you. (Can select more than 1)

☐ Stress ☐ Anxiety

☐ Depression ☐ Loss of job

☐ Social isolation ☐ Troubled relationships

Others: _____

Section C: Symptoms that matter most

Please tick ☑ where appropriate.

13. What specific activities that are most important to you/your loved one and you/your loved one not able to do because of SMA? (Select TOP 4)

- ☐ Independence in mobility (around the house, to work, to school)
- ☐ Feed oneself
- ☐ Ability to spend time alone / be independent
- ☐ Engage in social activities and building relationships (playdates, dining out, dating, hugging my partner)
- ☐ Attend work or school
- ☐ Engage in physical activities (playing sports, going to the gym)
- ☐ Transferring (from wheelchair / scooter to bed, toilet, etc.)
- ☐ Attend to personal hygiene independently
- ☐ Dress oneself
- ☐ Going to restroom by oneself
- ☐ Turning in bed
- ☐ Others:

_____

_____

_____

_____

_____

14. Which of the following symptoms currently has the most significant impact on you/your loved one's life? (Select TOP 4)

- ☐ Breathing difficulties (shallow, rapid, depressed breathing, etc.)
- ☐ Communication difficulties
- ☐ Inability to cough/clear lung secretions
- ☐ Feeding/swallowing difficulties
- ☐ Muscle weakness (facial, neck, arms, forearms, hips, legs)
- ☐ Breathing/lung infections (e.g. pneumonia, viral infections, etc.)
- ☐ Respiratory Failure requiring assistive devices (BiPAP, Ventilator, etc.)
- ☐ Joint contractures (tight muscles and tendons) / severe scoliosis
- ☐ Fatigue
- ☐ Falls
- ☐ Sleep problems
- ☐ Bone fractures/Hip dislocation
- ☐ Paralysis
- ☐ Others:

_____

_____

_____

_____

Section D: Management and treatment options for SMA

Please tick ☑ where appropriate.

15. In the last 12 months, how often have you/your loved one had to go to the hospital, for emergency care or inpatient hospitalization due to your/your loved one's SMA?

☐ None ☐ 1-2 times

☐ 3-5 times ☐ 6-10 times ☐ More than 10 times

16. In the last 12 months, how often have you/your loved one had to go to a doctor or medical specialist for routine care, or follow up of your/your loved one's SMA?

☐ None ☐ 1-2 times

☐ 3-5 times ☐ 6-10 times ☐ More than 10 times

17. Are you/your loved one visiting any of these healthcare professionals? Please state how many times a year. (Can select more than 1)

Healthcare Professionals	Times/year
☐ Neurologist	
☐ Geneticist	
☐ Respiratory specialist/ Pulmonologist	
☐ Orthopedic specialist	
☐ Mental health professional (psychiatrist, psychologist, counsellor)	
☐ Nutritionist/Dietician	
☐ Complimentary medicine specialist (Acupuncture, herbs, Ayurvedic, massage, homeopathy, etc.) Specify: _____	
☐ Others: _____	

18. Are you/your loved one receiving any of the services? Please state how many times a year.
(Can select more than 1)

Services	Times/year
☐ Geneticist	
☐ Physiotherapy	
☐ Occupational therapy	
☐ Hydrotherapy	
☐ Palliative care	
☐ Others: _____	

19. What is the estimated annual SMA related expenses/costs for you and your family?

	Cost/year (RM)
☐ Medical professional (doctor, nurse)	
☐ Allied health (Physiotherapist, speech therapist, occupational therapist, hydro therapist, psychologist, counselor, etc.)	
☐ Medicine (Over counter medicine, prescriptions, etc.)	
☐ Medical supplies (NG tubes, mask, suction catheter, gloves, etc.)	
☐ Nutritional support (supplementary diet, etc.)	
☐ Orthotics' support/adaptive devices (stander, splints, pencil holder, etc.)	
☐ Adaptive home renovation (ramps, rails, larger doors, etc.)	
☐ Mobility devices (wheelchair, crutches, scooters, walking frame, etc.)	
☐ Respiratory maintenance (cough assist machine, suction machine, BiPAP, etc.)	
☐ Travel & accommodation (SMA related)	

20. Have you/your loved one ever been prescribed (either by your doctor or through a trial) and taken the following medications? Select those which are applicable to you/your loved one.
(Can select more than 1)

☐ Albuterol (inhaled) ☐ Albuterol (liquid) ☐ Albuterol (tablet)
☐ Carnitine
☐ Creatine
☐ Hydroxyurea
☐ Steroids
☐ Valporoic Acid (VPA)
☐ Sodium Phenylbuterate
☐ Riluzole
☐ Nusinersen/Spinraza
☐ Others: _____

21. Which of these do you/your loved one use for respiratory assistance at home? Select those are applicable to you/your loved one. (Can select more than 1)

☐ Chest physiotherapy (CPT) for clearance/comfort
☐ Postural Drainage
☐ High frequency chest wall oscillation (VEST ©)
☐ Cough Assist Device
☐ Suction to remove secretions
☐ Non-invasive ventilation (NIV), such as BiPAP
☐ Invasive ventilation / Mechanical ventilator (with tracheotomy)
☐ Other: _____

22. Have you/your loved one undergone Scoliosis surgery (growing rods or spinal fusion)?

☐ Yes ☐ No

23. Are you aware of any drugs developed to treat SMA?

☐ Yes ☐ No

24. Which outcome below would you regard as the most important for a possible drug treatment? Select ONE option.

☐ The treatment will provide gains in function
(e.g., increased strength, energy, doing something I was unable to do before)

☐ The treatment will lessen symptoms that would improve my/my loved one's current quality of life and /or allow for enhanced activities of daily living

☐ The treatment will stop or slow down disease progression (even if does not provide lessening of symptoms that would improve my/my loved one's current quality of life and /or allow for enhanced activities of daily living)

☐ The treatment will prolong life span

☐ Other: _____

_____

25. Which of the following factors would influence your decision to not use or stop a given treatment? Select those are applicable to you. (Can select more than 1)

☐ The significant risks of serious side effects such as cardiac or kidney issues

☐ The common side effects of the treatment, such as nausea, loss of appetite, etc.

☐ The way that treatment is administered (for example, orally, intravenously, intrathecally),

☐ How long the treatment takes, whether it requires hospitalization, required doctors' visits, etc.

☐ The time that it would take away from my daily activities, job, school, etc.

☐ The burden of administration, such as the need for anesthesia, radiation exposure, surgical procedure, etc.

☐ Cost

☐ Other: _____

_____

26. Have you /your loved one ever participated in any type of clinical trial studying experimental treatments for SMA?

☐ Yes

☐ No. Please choose your reason(s):

☐ I am not aware of any clinical trial.

☐ Tried to enroll in a clinical trial, but did not qualify

☐ Tried to enroll but trial enrolment was closed

☐ Tried to enroll but no response

☐ Did not want to enroll due to burden of trial (travel, potential risks, time missed from work, etc.)

☐ Did not want to enroll for other reasons.

Please specify: _____

☐ I've received access to an experimental drug through an Expanded Access / Compassionate Use Program but did not participate in a clinical trial for this drug

☐ I'm not sure

27. Which of the following factors would you regard as the most important to your decision about whether to participate in a clinical trial to study an experimental treatment? Select TOP 4.

☐ Reputation of study site doctor

☐ Common side effects (headache, back-pain, skin rashes)

☐ The risk of rare but serious side effects (life threatening allergic reaction)

☐ How the treatment might prevent further disease progression or improve my/my loved one's health

☐ How the trial might affect my/my loved one's current treatment plan

☐ Promise of receiving open label therapy at the end of the study

☐ Proximity of the study site

☐ Frequency of visits

☐ Duration of visits

☐ Availability of safety data

28. Have you heard of preimplantation genetic diagnosis (PGD)?

☐ Yes ☐ No

29. Would you consider PGD service in future?

☐ Yes ☐ No

30. Have you heard of prenatal diagnosis (PND)?

☐ Yes ☐ No (Please proceed to question no. 35)

31. Have you undergone PND?

☐ Yes ☐ No (Please proceed to question no. 35)

32. What was the outcome of the PND?

☐ Fetus affected

☐ Fetus not affected

33. What was your decision if the fetus was affected?

☐ Continue pregnancy

☐ Termination of pregnancy

34. Were you satisfied with the PND service?

Not satisfied at all	Less satisfied	Neutral	Satisfied	Very Satisfied
☐	☐	☐	☐	☐
1	2	3	4	5

35. Would you consider PND service in future?

☐ Yes ☐ No

36. Have you heard of newborn screening?

☐ Yes ☐ No

37. Would you consider newborn screening in future?

☐ Yes ☐ No

Thank you very much for your participation. Your participation will help towards effort in improving the care and support for SMA in Malaysia, including access to medicines.

Are you willing to participate in a follow up discussion?

☐ Yes ☐ No

Appendix 2

Please read each statement and circle a number 0, 1, 2 or 3 which indicates how much the statement applied to you over the past week. There are no right or wrong answers. Do not spend too much time on any statement.

The rating scale is as follows:

0 = Did not apply to me at all
1 = Applied to me to some degree, or some of the time
2 = Applied to me to a considerable degree, or a good part of time
3 = Applied to me very much, or most of the time

1	I found it hard to wind down	0	1	2	3
2	I was aware of dryness of my mouth	0	1	2	3
3	I couldn't seem to experience any positive feeling at all	0	1	2	3
4	I experienced breathing difficulty (eg, excessively rapid breathing, breathlessness in the absence of physical exertion)	0	1	2	3
5	I found it difficult to work up the initiative to do things	0	1	2	3
6	I tended to over-react to situations	0	1	2	3
7	I experienced trembling (eg, in the hands)	0	1	2	3
8	I felt that I was using a lot of nervous energy	0	1	2	3
9	I was worried about situations in which I might panic and make a fool of myself	0	1	2	3
10	I felt that I had nothing to look forward to	0	1	2	3
11	I found myself getting agitated	0	1	2	3
12	I found it difficult to relax	0	1	2	3
13	I felt down-hearted and blue	0	1	2	3
14	I was intolerant of anything that kept me from getting on with what I was doing	0	1	2	3

15	I felt I was close to panic	0	1	2	3
16	I was unable to become enthusiastic about anything	0	1	2	3
17	I felt I wasn't worth much as a person	0	1	2	3
18	I felt that I was rather touchy	0	1	2	3
19	I was aware of the action of my heart in the absence of physical exertion (eg, sense of heart rate increase, heart missing a beat)	0	1	2	3
20	I felt scared without any good reason	0	1	2	3
21	I felt that life was meaningless	0	1	2	3

Appendix 3

The impact of living with spinal muscular atrophy in Malaysia from Patients and Caregivers perspectives (MySMA)

Interview Guides for Qualitative Research

A. Experience related to diagnosis

1. Can you share your experience on how your/your love one's diagnosis was made? What was the first symptom?

2. How do you feel after you/your love one's are diagnosed?

3. Did the doctor explain the condition of SMA to you?

B. Impact on life

4. How does SMA affect your/your love one's life?
   a. Intrapersonal (How you perceive yourself, your feelings)
   b. Interpersonal (Relationship with your family, relatives,friends,colleagues etc)

C. Worries and Concern

5. What are some of the things you are most worried about?
   a. Confronting premature death
   b. Difficult treatment choices
   c. Heartbreak and fear with loss of functional abilities
   d. Coming to terms with lost expectations
   e. Loss of sleep and stress
   f. Social discomfiture and stigma
   g. Limitations on social activities
   h. Struggle to achieve Independence
   i. Uncertainty and helplessness
   j. Pressure on family finances

6. How big of an impact has this item had on [your life/the life of the person you care for]? (Prompt: for each item that has been mentioned by participants/caregiver, ask them to rate each on a scale of 1-10 to their life.)

- eg: Client 1: client mentioned that social stigma and finance is the main issues, then:
- Facilitator asks the client: In a scale of 1-10, please rate the impact of how financial issue has on to your life.
- Or prepare a piece of paper (likert scale template) to be used by researchers to ask participants (to rank from 1-10)

D. Hopes and concerns for Treatment

7. [If not mentioned] do [you/the person you care for] do things/take steps to avoid being hospitalized? Tell me about that?

8. Do [you/the person you care for] do things/take steps to avoid [respiratory events/things that might impact breathing]? Tell me about that?

9. In the past 12 months, what kind of decline in motor function did [you/the person you care for] experience?

10. Would a treatment that stops decline in motor function be meaningful to [you/the person you care for]?

11. Would a treatment that improves your motor function be meaningful to [you/the person you care for]?

12. What kind of improvement do [you/the person you care for] want to see from treatment?

Living with SMA. #thinksmallactbig.

IN LOVING **MEMORY**

All the SMA ANGELS
Forever cherished
in our hearts.

JAMES LOW,
Departed
27.5.2020
Lawyer,
disability rights activist
and SMA patient

Your strength,
your resilience,
your joy,
your faith,
and
your attitude of
living life to the fullest
serves as
a shining beacon for
the SMA community.
Your legacy will live on forever.