Original Article

The Difficulties and Coping Experiences of Families of Children with Spinal Muscular Atrophy, towards the Diagnosis and Treatment Process: A Phenomenological Study

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Abstract

Objective: The aim of this study was to evaluate the difficulties and coping experiences of families with children with SMA (spinal muscular atrophy) in using a phenomenological approach.

Material-Method: This qualitative study was conducted with semi-structured in-depth interviews via whatsapp with 35 parents with children with SMA (spinal muscular atrophy) living in Turkey. Interviews continued until data saturation was achieved. All interviews were audio recorded and then transcribed. The data were analyzed using Colaizzi's phenomenological analysis method. The study was conducted and reported according to the COREQ checklist.

Findings: Data analysis revealed two themes (psychosocial problems experienced and difficulties and coping experiences of parents) and five sub-themes (mental, social, physical, difficulties and coping).

Conclusion: It was determined that parents experienced many different psychosocial problems due to SMA disease. In addition, it has been determined that parents are partially or completely isolated from the society due to the burden of care, they experience disruptions in communicating with their family and close circles, and their lives are negatively affected.

Keywords: Turkey, SMA patients, coping, parents, phenomenological study

Introduction

Spinal Muscular Atrophy (SMA) is an inherited muscle disease. The main reason why SMA occurs in people is due to mutations in the SMA1 gene. Genetic problems in the SMA1 gene cause the SMN protein not to be produced, and if the SMN protein is not produced, motor neuron cells shrink and die (Gulec, 2021). Although the symptoms of SMA disease vary from individual to individual, patients may experience lying in the frog leg position, involuntary contraction of the tongue (fasciculation), weak muscle structure, arm and leg behaviors, lost deep tendon reflexes, weakness, inability to walk, problems in the respiratory system, nutritional disorders, weak crying and lagging behind their peers (Turkey SMA Foundation, 2017, Haspolat and Koken, 2022). On the other hand, when we look at the mortality rates in SMA patients, this disease appears as one of the biggest causes of infant mortality. It is seen that mortality rates in SMA patients are higher compared to the recovery rate. The most important factor in the increase in mortality rates is the lack of access to treatment facilities and necessary medications. One of the main problems experienced at the point of access to medicines is that individuals with SMA and their families cannot afford the very high cost of medication and health policies are not structured in a way to fully take into account the needs of SMA patients (Urek and Karaman, 2019).

SMA patients and their families have many physical, psychological, social and economic problems and needs (Akgun Kostak & Cetintas, 2022). Especially the diagnosis process is a very troublesome process in itself and can shape the treatment process. In addition, many psychosocial problems are experienced during the treatment process (Kruitwagen-Van Reenen et al., 2016, Catulay et al., 2023).

Chronic diseases cause devastating psychosocial problems for individuals and their families (Aydemir & Cetin, 2019). Since SMA is a rare disease with high physical difficulties and limited access to treatment tools, individuals and their families struggling with this disease experience psychosocial problems (Aslanturk, Derin & Arslan, 2019). Emotional problems are at the top of these problems. Facing a diagnosis of a chronic disease such as SMA can lead to a range of emotions such as sadness, anger, helplessness, frustration and anxiety. However, sometimes there are serious difficulties in the process of accepting the disease. Individuals diagnosed with SMA or their families may tend to reject the diagnosis as a 'punishment' or 'sin'. This may lead to less and difficult adaptation to the treatment process (Kruitwagen-Van Reenen et al., 2016; Catulay et al., 2023). In this context, the study aims to evaluate the difficulties and coping experiences of families with children with SMA (spinal muscular atrophy) regarding the diagnosis and treatment process using a phenomenological approach.

Method : Throughout this study, the author followed the Consolidated Criteria for Reporting Qualitative Research (COREQ) and reported accordingly (Tong et al., 2007).

Table 1. Combined criteria for reporting qualitative research (COREQ).

Area 1: Research team and reflexivity

Personal Characteristics

Number	Item	Guiding questions	Explanations
1	Interviewer/facilitator	Which author/authors conducted the interview or focus group?	The second author conducted the interview.
2	Identity information	What were the credentials of the researcher, e.g. PhD, MD	First author PhD Second author Bachelor's degree nurse
3	Profession	What was their occupation at the time of the study?	First author: Dr. Faculty Member, Psychiatric Nursing Second author Nurse
4	Gender	Was the researcher a man or a woman?	Two researchers Female
5	Experience and training	What experience or training did the researcher have?	The first author has taken qualitative courses, has experience in qualitative research and has published qualitative studies in international journals.

Relationship with participants

6	Relationship status	Was a relationship established before the training started?	No relationship was established before the start of the study.
7	Interviewer's participant knowledge	What did participants know about the researcher, e.g. personal goals, reasons for doing research?	Parents knew that the researcher worked as a nurse
8	Interviewer characteristics	What characteristics were reported about the interviewer/facilitator, e.g. bias, assumptions, motives and interests in the research?	At the beginning of each interview, parents were informed about the purpose and objectives of the study.

Area 2. Study design

Theoretical framework

9	Methodological orientation and Theory	Which methodological orientation was specified to support the study, e.g. discourse analysis, ethnography, phenomenology, content analysis?	This was a phenomenological study.

Participant selection

10	Sampling	How were participants selected? e.g., purposeful, convenience, consecutive, snowball	Criterion sampling method, one of the purposive sampling methods, was used.
11	Approach method	How were participants approached, e.g. face-to-face, telephone, mail	The timing of the interviews was determined by the individuals who voluntarily agreed to participate in the study.
12	Sample size	How many participants were there in the study?	A total of 35 parents were in the study.
13	Disagree	How many people refused to participate or dropped out? Reasons?	There were no individuals who refused to participate in the study.

Setting

14	The setting of data collection	Where was the data collected? e.g. home, clinic, workplace	Detailed information is provided in the data collection section of the study.
15	Presence of non-participants	Was there anyone else present apart from the participants and the researchers?	There were no observers.
16	Description of the sample	What are the important characteristics of the sample, e.g. demographic data, history	Nurses who agreed to participate in the study were included in the study.

Data collection

17	Interview guide	Were questions, prompts and guidelines provided by the authors? Has it been pilot tested?	Detailed information was given in the Methods section.
18	Repeat interviews	Have there been re-interviews? If yes, how many?	No.
19	Audio/visual recording	Was audio or visual recording used to collect data in the study?	Interviews were recorded with a voice recorder
20	Field notes	Were field notes taken during and/or after the interview or focus group?	All nurses' responses and researcher observations were recorded.
21	Duration	How long were the interviews or focus groups?	Each interview lasted between 35 and 45 minutes.
22	Data saturation	Has data saturation been discussed?	Data saturation was discussed.
23	Transcripts returned	Have transcripts been returned to participants for comments and/or corrections?	No.

Area 3: analysis and findings

24	Number of data coders	How many data coders coded the data?	Two researchers and a third individual coded the data.
25	Description of the coding tree	Did the authors provide a description of the coding tree?	The titles and subtitles in the results section represent the final coding tree.
26	Derivation of themes	Were the themes predetermined or derived from the data?	Themes were derived from the data.
27	Software	What software, if any, was used to manage the data?	Data were analyzed manually.
28	Participant control	Did participants provide feedback on the findings?	No.

Reporting

29	Quotes provided	Are participant quotes presented to illustrate themes/findings? Is each quote identified, e.g. participant number	Source. Participant quotes are provided to illustrate themes/findings. e.g. participant number.
30	Data and findings consistent	Was there consistency between the data presented and the findings?	Yes
31	Clarity of main themes	Are the main themes clearly presented in the findings?	Yes
32	Clarity of small themes	Is there an explanation of the different cases or	Yes

		a discussion of minor issues?	

Study Design: This study was conducted between November 2023 and December 2023 using an inductive qualitative design. It was conducted through semi-structured in-depth interviews conducted via whatsapp with 35 parents who live in Turkey and have a child with SMA (spinal muscular atrophy).

Research team and reflexivity: The first researcher was working as an active faculty member (doctoral faculty member) in a nursing school. The researcher has a doctorate in psychiatric nursing. She also had experience working as a clinical nurse and was trained in qualitative research methods. The second researcher was working as an intensive care nurse with the participants in a hospital in the south of the country.

Working group: Criterion sampling method, which is one of the purposeful sampling methods, was used to determine the study group of the research. Criterion sampling is the sampling of people, events, objects or situations that have the qualities determined for the problem (Altunay et al., 2014; Baskaya & Demir, 2023). Semi-structured in-depth interviews were conducted with 35 parents who live in Turkey and have a child with SMA (spinal muscular atrophy) via whatsapp and were terminated when data saturation was reached.

The inclusion criteria were as follows: (a) having a child with SMA (spinal muscular atrophy), (b) being open to communication, and (c) agreeing to participate in the study. The exclusion criteria were as follows: (a) having a speech, language or hearing impairment that would prevent communication and (b) not agreeing to participate in the study.

Data collection: A semi-structured interview form was prepared by the researchers based on the related literature review. The form consists of two parts. The first part includes 5 questions about the age, gender, education and economic status of the parents, the age of the child and the age of diagnosis. The second part consists of a form with basic open-ended questions to be used in the semi-structured interview. The questions in the semi-structured interview form were conducted through semi-structured in-depth interviews via whatsapp (Brinkmann & Kvale, 2015). In the interviews, parents were asked questions about the difficulties they experienced during the diagnosis and treatment process and were asked to describe their coping experiences and asked questions such as "Can you elaborate on your answer?" and "What do you mean by this?" All interviews were conducted by (second author). The interviews were recorded using a voice recorder and transcribed verbatim by the researchers. After all interviews were completed, the study data were transcribed for analysis.

Questions in the semi-structured interview form;

1.What does the word SMA mean to you?
2.What do you think about the SMA diagnosis process?
3.What do parents with a child diagnosed with SMA experience? What are the psychological, social and physical effects on you?
4.Do you think you are coping effectively with the SMA treatment process? Which difficulties do you experience?
5.What symptoms is your child experiencing? What are you experiencing to cope with these symptoms?
6.What would you like to see done for you?

Data analysis: In the analysis of the qualitative data obtained from the interviews, the 7-step analysis method developed by Colaizzi (1978) for phenomenological studies was used (Morrow et al., 2015). In this context, the interview texts were read by the researchers. Thus, it was tried to understand what was explained in the data. Important statements in the interview texts were selected, reorganized and expressed in general terms. Then, the data that were tried to be explained in the statements were identified and analyzed. The researchers then identified and organized the themes into main and sub-themes. The themes and sub-themes of the study were developed through clear expression. In addition, by including the statements of the participants, the reader was able to verify the interpretation and analysis of the data (Yildirim et al., 2021; Ulutasdemir et al., 2022)

Ethical Aspects of the Research: This study was approved by Gümüşhane University Scientific Research and Publication Ethics Committee (E-95674917-108.99-181916). Informed consent was obtained from the participants before starting the interview. Recordings and transcripts were stored on a password-protected device. The study was conducted in accordance with the Declaration of Helsinki and the ethical standards of the National Research Committee.

Findings

The mean age of the parents included in the study was 28.57 ± 4.8; twenty-eight of them were women and most of them had poor economic status. The mean age of children with SMA was calculated as 2.52± 0.52. Demographic characteristics of the individuals participating in the study are presented in Table 2.

As a result of the analysis of the data obtained from the semi-structured interviews, themes, sub-themes and codes were identified (Table 3).

Table 2. Characteristics of Participants

Participant number	Age	Gender	Age of the child	Child's age at diagnosis	Economic situation
K1	24	Woman	2 years old	5 months	Bad level
K2	33	Woman	1 year old	10 months	Medium level
K3	26	Male	1.5 years old	4 months	Medium level
K4	27	Woman	1 year old	3 months	Medium level
K5	22	Woman	2.5 years old	1 month	Bad level
K6	24	Woman	2 years old	5 months	Medium level
K7	24	Woman	1.5 years old	2 months	Medium level
K8	37	Male	1 year old	7 months	Medium level
K9	27	Woman	1 year old	2 months	Bad level
K10	34	Woman	1.5 years old	4 months	Bad level
K11	32	Woman	1 year old	11 months	Bad level
K12	32	Woman	2.5 years old	10 months	Bad level
K13	38	Woman	2 years old	8 months	Bad level
K14	26	Woman	1.5 years old	5 months	Medium level
K15	25	Woman	1 year old	6 months	Bad level
K16	25	Male	1 year old	5 months	Medium level
K17	26	Male	1.5 years old	10 months	Medium level
K18	27	Male	1 year old	4 months	Bad level
K19	22	Woman	2 years old	3 months	Bad level

K20	24	Woman	1 year old	40 days	Bad level
K21	24	Woman	2.5 years old	1 month	Bad level
K22	37	Woman	2 years old	4 months	Bad level
K23	27	Woman	1.5 years old	3 months	Medium level
K24	34	Woman	1 year old	40 days	Bad level
K25	32	Woman	1.5 years old	3 months	Medium level
K26	32	Male	1 year old	5 months	Bad level
K27	28	Woman	2 years old	10 months	Medium level
K28	22	Woman	2.5 years old	4 months	Medium level
K29	24	Woman	2 years old	3 months	Bad level
K30	24	Woman	1.5 years old	40 days	Medium level
K31	37	Woman	1 year old	1 month	Medium level
K32	27	Woman	1 year old	3 months	Bad level
K33	34	Woman	2 years old	1 month	Bad level
K34	32	Male	1.5 years old	4 months	Medium level
K35	31	Woman	1 year old	3 months	Medium level

Table 3. Difficulties and Coping Experiences of Families with Children with SMA (Spinal Muscular Atrophy) Related to the Diagnosis and Treatment Process

Themes	**Sub Themes**	**Codes**
1. Psychosocial problems experienced	A. Social	A1. Social isolation
		A2. Weakening of peer relations
		A3. Stigmatization
		A4. Lack of interest in aid campaigns
	B. Spiritual	B1. Collapse
		B2. Burnout
		B3. Fear of loss
		B4. Anxiety
		B5. Future anxiety
		B6. Despair
		B7. Shock
		B8. Denial
	C.Physical	C1. Fatigue
		C2. Insomnia

		C3.Economic problems C4. Not working at work C5. High maintenance burden
2. Challenges and coping experiences of parents	A. Challenges faced	A1. Economic challenges A2. Insufficient support from the government A3. The high cost of medicines because they are imported from abroad A4. Affecting social environment relations A5. Family problems A6. Lack of psychological support
	B. Coping	B1. Prayer B2. Supporting hope B3. Organizing aid campaigns B4. Self-indulgence B5. To do spiritual self-care B6. Reading a book B7. Seek refuge in God B8. Being patient B9. Praying

Theme 1. Psychosocial problems experienced

Subtheme 1. Social

According to the data obtained from the interviews, parents experience social problems such as social isolation, weakening of peer relations, stigmatization and lack of interest in aid campaigns.

> *"We had a lot of difficulties in the hospitals in Istanbul, my son bruised in our hands many times, we are very scared, our only hope is that you get the medicine abroad, we are very tired of this process, we are very worn out in hospitals. Anyway, your relations with your relatives are over and you don't see anyone... (P7)*

> *""We have completely reduced our social life to zero. We live an isolated life. Our family elders cannot come to our house due to infection, and even if they do, they see our daughter from a very short distance in a sterile way and leave." (P6)*

Subtheme 2. Spiritual

According to the data obtained from the interviews, it was found that parents were also negatively affected mentally.

> *"You feel helpless, you are mentally very bad, you smile at your baby and play games, but inside you are rotting, there is no social life at all, I return from work early and try to help my wife in the care of our daughter, of course, my wife is also very difficult as a mother, it is really a very difficult situation." (P26).*

> *"Families with a child with SMA experience helplessness, they experience economic difficulties, they may even want to break their own lives, you are not really well mentally, you are not really well mentally, because this mental state is not good, it is not good with your body, of course, this process is spent only at home trying to make your baby's day better than the other day, really,*

> *sometimes people can even get depressed because they are helpless... (P12).*

Subtheme 3. Physical

According to the data obtained from the interviews, it was found that parents were also physically affected in a very negative way.

> *"People cannot get anywhere financially, you say that your money is enough for us, but we are trying to collect money for the treatment of your child and unfortunately we are collecting, we are very bad mentally, our child is melting in front of our eyes, in general, we make a lot of effort, but it is quite difficult, and there is no such thing as social life, we cannot meet with friends, we cannot meet with friends, we usually have to withdraw ourselves from social life in this process because we are interested in the care and treatment of our child." (P32).*

> *"This is a very difficult disease. Fatigue, insomnia, aggressive behavior.... My husband can go out, but sometimes I can take all my anger out on him because I cannot go out. When he tells me to go out, I can't because I can't leave my child. I actually love going out, but when I leave my child, I can't go out because my mind will stay with him." (P21)*

Theme 2. Challenges and coping experiences of parents

Subtheme 1. Difficulties experienced

Parents stated that they had a lot of difficulties due to their children having SMA. In particular, parents experience many different problems such as economic difficulties, inadequate support from the state, the high cost of medicines due to the fact that they come from abroad, affecting social relations and domestic problems.

> *"These children need special devices. While the state provides around 20% support to cover these devices, we have to cover the rest ourselves as families. The cost of these devices is very high, so it is a disease with a great economic burden. Unfortunately, we cannot get our treatment." (P12)*

> *"These children receive very intensive physical therapy. Apart from the physical therapy covered by the state, I pay for it privately on the other days and this fee is 3500 TL. Imagine that every week you spend 3500 TL only for physical therapy...This puts a big burden on your back economically." (P29)*

Subtheme 2. Coping

While caring for their children, parents reported that they cope by promoting hope, practicing spiritual self-care, reading books, praying and being patient.

> *"No one talked to anyone at home. No laughing, no talking, no fun. Our focus was only on our daughter. Of course, this miscommunication was affecting our psychology more and more day by day. Almost all of us collapsed mentally." (P19)*

> *"Sometimes, as a father, I cannot keep up with some things and this is a very sad situation for me, thanks to the volunteers, they are very supportive, you are in a foreign city, you are going to go to the hospital, there is no road, no trace, we call the volunteers, they help us, they make appointments, they help us buy the device, I pray to my God, I try to cope, but believe me, it is a really difficult situation... (P23).*

Discussion

The aim of this study was to evaluate the difficulties and coping experiences of families with children with SMA (spinal muscular atrophy) regarding the diagnosis and treatment process using a phenomenological approach. The difficulties and coping experiences of families with children with SMA (spinal muscular atrophy) regarding the diagnosis and treatment process were discussed in two parts.

Psychosocial problems experienced

In the study, it was determined that families with children with SMA experienced many psychosocial problems during the diagnosis and treatment process. Parents stated that they experienced social isolation and stigmatization. In the study conducted by Bekiroglu and Gokcearslan Cifci (2017) on the psychological status of families of neuromuscular patients, it was found that only 36.5 percent of the families evaluated their current mental health as good/very good, the vast majority did not see themselves as good and cheerful (91%) and almost two-thirds felt tired (Bekiroglu & Gokcearslan Cifci, 2017).

Parents stated that they were very mentally exhausted and experienced depression and helplessness. They also stated that they experienced situations such as shock and denial when they first encountered the disease. Kutlu and Acikgoz (2023), in their study with caregivers of individuals with Spinal Muscular Atrophy, asked the participants about their reactions after the first encounter with the disease and stated that they were in a situation such as shock, denial, fear, and not knowing what to do (Kutlu & Acikgoz, 2023). In Korkut Orhan's (2019) study, it was stated that the first reactions of nine parents of children with SMA to the disease were feelings such as shock/confusion, sadness, destruction, and fear. At this point, it can be said that the disease has created common feelings and thoughts in people.

Parents with children with SMA stated that they also experienced physical problems such as fatigue and insomnia. Ergenekon et al., (2023) found that depression, trait anxiety and insomnia were common in SMA caregivers (Ergenekon et al., 2023). All these results can be accepted as an indicator that SMA disease affects individuals physically, socially and spiritually in many ways.

Challenges and coping experiences of parents

As a result of the study, it was determined that parents had great difficulty in coping with many problems they experienced. In the study conducted by Evkaya Acar et al. on the burden of SMA caregivers, a relationship was found between the income level of families and their needs (Evkaya Acar et al., 2021). In another quantitative study in our country, it was found that depression, trait anxiety and insomnia were common in SMA caregivers (Ergenekon et al., 2023). All these results can be said that SMA disease affects individuals in many ways .

In a study conducted in Canada, the situation of both SMA patients and their caregivers was examined. In the study, it was determined that the quality of life of the individuals was affected, problems in work-school attendance, various health problems emerged in caregivers, and new costs emerged due to travel for treatment purposes and devices (McMillan et al., 2021). In another study, it was reported that parents of children with SMA are overburdened (Martínez-Jalilie et al. 2020). In another study conducted with 219 relatives of patients with rare diseases, 25 of whom had SMA, it was determined that 74.4 percent of family members needed psychosocial help and support. In other words, relatives of patients consider such support to be necessary to a great extent (Pak Gure, 2021). It was concluded that patient relatives are mentally worn out due to the disease and that they need to be psychologically strengthened.

Conclusion: Since the diagnosis of SMA, there have been significant social, psychological and economic changes in the lives of caregivers. It has been determined that parents are partially or completely isolated from the society due to the burden of care, they experience disruptions in communicating with their family and close circles, and their lives are negatively affected. It has been determined that individuals experience intense sadness, fear and fear of loss during the disease process, families have difficulties in meeting these costs due to the high cost of medical products and devices, and that they restructure their entire lives with the introduction of this disease into their lives

In line with the results of the study, it is thought that caregivers should be supported psychosocially. Health policies and social policies are of great importance in supporting individuals and families with SMA. In this context, it is recommended to develop policies, services, programs and legal regulations that address the needs in their entirety and support practices to meet them.

Limitations: One of the limitations of the study is that all participants were selected from parents with SMA living in Turkey. The results depend on the participants and the setting in which the research was conducted. The small group of participants does not represent the entire world population of children with SMA.

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