

# The lived experiences of patients with spinal muscular atrophy: A phenomenological study

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## Abstract

**Context:** Spinal muscular atrophy (SMA) is a neuromuscular disease which, in addition to physical disability, causes psychological and social problems for patients and their families. If physiological and mental disorders continue to persist for a long time, it leads to serious deterioration in the patient's physical condition, influences the treatment process, and prevents the outcome of the therapies.

**Aims:** The aim of this study was to explore the lived experiences of patients with SMA.

**Setting and Design:** In this qualitative study, a total of six participants (five patients and one patients' wife) were selected by purposive sampling method.

**Material and Methods:** Data were collected by semi-structured interview. Eleven interviews were conducted and analyzed by Colaizzi's descriptive phenomenological approach.

**Statistical Analysis Used:** Data were analyzed by One Note software version 14.0 with Colaizzi's descriptive phenomenological approach.

**Results:** Two main themes – “tragic life” and “vague future” – emerged from data analysis. They had four categories including “physical and psychological problems,” “limitations and lost opportunities,” “concerns,” and “uncertain future.”

**Conclusions:** These patients experienced a lot of physical and psychological impairments in their life. They have many concerns and lost some opportunities. Therefore, policymakers and authorities should support these patients for participating as an effective members in the society, consequently their feeling improve by getting independent.

**Keywords:** Lived experiences, Qualitative research, Spinal muscular atrophy

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## INTRODUCTION

Spinal muscular atrophy (SMA) is a motor neuron degenerative disease, which is the most common autosomal recessive disorder after cystic fibrosis.<sup>[1]</sup> In this case, motor neurons of the anterior horn cells are degenerated. Moreover, the patient encounters disability in

some forms of voluntary movement.<sup>[2]</sup> The probability of being carriers is 1.40, and the incidence of this disease is 1 in 6000 to 10,000.<sup>[3]</sup> Clinically, it has three different types. In type one, the most severe type of illness, the age of the symptoms is from birth to 6 months. Embryos with this disorder have relatively small movements. In this type of illness, the baby is unable to swallow and sit down and face

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difficulty in sucking and breathing. They usually die from respiratory problems before the age of 2 years.<sup>[4]</sup> In the second type of disease, the age of the onset of symptoms is from 6 months to 2 years. These patients can sit alone, but have difficulty in walking. Due to the severity of the condition, most patients live no longer than 10 years. The third type of disease is known as Kugelberg–Welander syndrome which is the mildest form of the disease. The onset of clinical symptoms is from 18 months and patients can walk.<sup>[2]</sup> A 10-year study in Saudi Arabia showed that 69% of relatives involvement reported in 14 cases of brother or sister.<sup>[5]</sup> Araghian *et al.* conducted a descriptive phenomenological study to explore childbirth experiences of a mother who had two children with Werdnig–Hoffman syndrome. The study findings indicated themes such as “family breakdown, gradual child death, child perception, and care for the patient’s child.”<sup>[6]</sup> In these patients, in addition to physical disabilities, psychological and social problems which originate from the nature and course of the disease occur for patients and their families, Low level of recreational activities in the family due to child’s illness and disruption of homework are the most common social problems.<sup>[7]</sup> Patients in addition to physical and mental problems suffer from economic poverty.<sup>[8]</sup> Based on clinical nursing experiences, patients with SMA encounter barriers which they must cope with. Health-care providers should have an understanding of such patients’ inner feelings, thereby providing appropriate care which is crucial in these patients.<sup>[9]</sup> Knowing the experience of patients with SMA is important to understand the concept of caring for chronic disability in such patients. It improves the health and social consequences of the individuals. Close nurse and patient communication helps to reduce the anxiety of these patients. Therefore, it is important to know the problems and concerns of patients and their feelings about the disease. Phenomenology gives researchers opportunity to understand the innermost aspect of the “lived experiences” of participants. As an approach that is “participant oriented,” phenomenology allows the participants to express themselves and their “lived experience” stories.<sup>[10]</sup> There is not clear understanding about the challenges of patients with SMA. The aim of this qualitative study was to explore the lived experience of patients with SMA.

## MATERIALS AND METHODS

### Design

This study explored the lived experience of patients living with SMA, by phenomenological approach, attempting to capture the essence of their lived experience. The data were gathered through interactive interviews. The audio

records of patients’ narrations of their experiences were transcribed verbatim. Probing questions were used to deepen the interviews.

### Participants and setting

The first participant was the researcher’s friend, and his sister was the second one. The purposive and snowball sampling methods were used to invite participants for interviews. Interviews were held at the participants’ home in Bojnord (north of Khorasan province, Iran). Inclusion criteria included individuals with confirmed diagnosis of SMA by a neurologist, being 18 years or older, being able to communicate, and willing to share their experiences. A total of 11 in-depth interviews with six participants (five patients’ SMA type III and one patient’s wife) were held between January 2018 and August 2018. Because one of the participants mentioned that her feeling and experiences changed by her marriage and her husband has a significant role in her feeling, we invited her husband to interview to enrich our research. Interviews lasted 30–45 min with respect to participants’ tolerance. Data saturation was reached by the sixth participant.

### Data collection

The participants were asked to describe their experience of living with SMA and their perceptions of life events from childhood to adulthood.

### Data analysis

All interviews were recorded by a voice recorder and listened by the researcher many times for deep involvement. Interviews transcribed word by word by the researcher, and the Colaizzi’s phenomenological approach by One Note software version 14.0 was used for data analysis.

(1) Each transcript was read and reread in order to obtain a general sense about the whole content; (2) for each transcript, significant statements that belong to the SMA experience were extracted; (3) meanings were formulated from the significant statements; (4) the formulated meanings were sorted into categories, clusters, and themes; (5) the findings of the study were integrated to enrich descriptions of the phenomenon of SMA; (6) the fundamental structure of SMA was described; and (7) confirmation of the results was obtained from the research participants to compare the investigator’s descriptive results with the experiences of participants.<sup>[11]</sup>

### Rigor

The rigor of this study was based on Lincoln and Guba’s evaluative criteria. For the credibility of the study, the analyst triangulation and member checking were used.

Data coding and analysis were done individually as well as collectively and shared with two expert researchers in qualitative research. Prior to the initiation of the study, two pilot interviews were conducted and checked by the researchers, and their comments were used to modify the subsequent interviews. For the member checking, data and conclusions were verified with the participants. For the transferability, the participants' descriptions of their life experiences were accounted. For fulfillment of dependability criteria, a researcher as an external audit who was not involved in the study process examined both the process and results of the research study. For the confirmability, the research advisor and one of the colleagues were invited to validate the data analysis.<sup>[12]</sup>

**Ethical consideration**

All ethical research codes, including the permission to record audio for interview; the confidentiality of individual identity; and the right to withdraw from the study were considered. All participants received required information about the purpose, method, and ethical rights, and informed consent form was completed. The study was approved by the Research Deputy and Research Ethics Committee of Golestan University of Medical Sciences (Project number: 960718188, Ethics Code: Ir.goums.rec. 1396.187).

**RESULTS**

Six participants (five patients and one patient's wife) participated in this study. Table 1 shows the detailed characteristics of the participants. Two main themes – “tragic life” with subthemes “Physical and psychological problems” and “Limitations and lost opportunities” and main theme “vague future” with two subthemes “concerns” and “uncertain prospect” – were extracted [Table 2].

**Table 1: The detailed characteristics of the participants**

Participants	Age	Gender	Education
Patient 1	27	Male	Bsc
Patient 5	32	Female	Bsc
Patient 2	29	Female	Bsc
Patient 4	26	Male	Associated degree
Patient 3	23	Female	Bsc
Patient's wife	31	Male	Bsc

**Table 2: Main themes and categories extracted from data analysis**

Theme	Categories
Tragic life	Physical and psychological problems Limitations and lost opportunities
Vague future	Concerns Uncertain prospect

**Tragic life**

By looking at the lives of these patients, it can be perceived that they go through difficult days, suffer from tremendous physical and psychological impairments, as well as the loss of social and occupational opportunities, marriage, and making a family.

**Physical and psychological problems**

According to Gayman et al. (2008), physical limitations are defined as a situation that restricts a person's capability to meet their social, familial, and occupational roles. Activity limitations are defined as difficulties in doing a task or action.<sup>[13]</sup> In this study, most participants reported the symptoms of weakness such as gaiting, sitting and getting up, climbing stairs, and running at the beginning of the disease. One of the participants stated that:

*“When I found out my disease that felt I could not walk, get up, run, or climb the stairs like the others. This feeling was more common in the elementary period, but from the age of six I felt being different.”* [P1].

Experience of participants from childhood indicates to feel of being different, visiting by many physicians, and hearing disappointed responses.

*“My parents did not know my illness because of illiteracy. Since childhood, they took me to many doctors, and they just said it is a congenital and incurable condition. My gait was different from the rest of the people, and I always dropped behind in school way. Sometimes I fell down.”* [P5].

With the exacerbation of symptoms, gradually, participants were not able to accompany their friends and peers during the school period. One of the participants said:

*“I do not have any good memories at all. I have no memory. My friends were playing and I had to watch them in a corner. They didn't counted me at all.”* [P5].

Physical disability was getting worse with the passage of time. The majority of participants could not able to meet their needs in their personal affairs at present and depend on their family members.

*“When I wake up, I have to call someone from the family to come and take my hands and helped me to get out of bed.”* [P1].

The experiences showed that they could not even meet their simple affairs:

*“My mum or sister carried me from courtyard to the taxi, then I will go to work. After returning home, they carry me down the stairs.”* [P5].

None of the participants had a good sense about their illness. Among participants, shame from being looked by others was a common feeling, for example, one of them said: *“Well, it did not feel good, because everyone looked at me and chattered their friends. I tried not to pay attention and continue my way”* [P1]. Other dominant feelings among participants were sorrow, feeling guilty of throwing family members in trouble, feel that their life is getting darker day by day, glooming, and regret of being born. One of them said:

*“It’s really hard and painful to lose my strength every day, and I can do nothing but look at my life that gets darker day by day.”* [P3].

Most participants tried to hide their problems from the others. One of them said:

*“When I think, I have never went to blackboard in guiding school. I talked my teacher not send me to the blackboard. I really liked to take chalk in my hand. I Remember that once I take chalk from school, I went home and wrote my homework on wall. At university even I talked to my professors. It was very hard for me that my classmates find out my disease.”* [P2].

The participants have expressed that they found themselves sometimes in a state of complete anguish. For example, one of them said: *“several times did I commit suicide during my adolescence. My mood was low. I always complained to God. Why me? among my siblings why me?”* [P5].

Although many participants disclose their illness as a life burden subject, in the first interview within some participants, they were trying to relieve themselves by comparing their conditions with the worst patients. For example, the participant number two said: *“In the beginning I asked God, why me? why my brother?..... but now that I get older, I say, my God, thank you for not getting worse, thank God, we breathe so easily and live easily. Maybe I’m getting wiser now.”* Occasionally, participants consider their illness as fate and divine wisdom and they are trying to relieve themselves. For example, one of them said: *“I accept my fate .... a leaf does not fall without the will of God”* [P5], but in a second interview with these participants, they showed no clue of coping and were involved with mental disturbances.

### Limitations and lost opportunities

After the onset of the disease, gradually, in addition to physical and psychological problems, they suffer from a lot of restrictions on their lives and have lost many opportunities. For example, one of them said: *“For the sake of problems, I was forced to choose an uninterested major in university. I couldn’t continue my education because of students strange looking,*

*stairs, and my disability for going. I love driving but I cannot have certificate because of my wrists’ and fingers’ weakness, I had some offers for job but I wasn’t able to accept them. I stay home because of this illness.”* [P1].

Some participants described numerous examples hardships for their inability in everyday activities:

*“I can’t join friends’ parties, unless I knew they have a sofa. because I was worried about sitting and standing up. I would not go outside with my friends because they walked steeply and there are places where I could not go.”* [P2].

Another participant said: *“I love winter ...no body knows how much I love this season. but I afraid so much, I remember I had so many problems for walking because of sleepy passage.”*

The patients lost many opportunities for hobbies, entertainment, and learning in their life. One of them said: *“I had several times liked to go to the classroom of music and language, but the places that I trained had a lot of stairs and were not suitable. The cost of a taxi was high. That’s why I gave up.”* [P3].

### Vague future

The story of the patients’ life illustrated that they have little hope for the future, see it dark and ambiguous, unable to plan, and this leads to feel helplessness and suffer.

### Concerns

The patients have numerous concerns about the future of the disease and its treatment, occupation, continuing education, marriage, and childbearing. For example, one of the participants was worrying about his dependency on her sister: *“If my parents get old, how can they take care of both of us? It’s always my mental concern, and I hate disturbed my older sister’s life. She can’t take care of Me and my sister.”* [P3].

Another participant talked about his concerns for finding a job with his condition: *“All my concern is finding a job...., does anyone ever accept me for work? Can I go over the task that give to me or not?...”* [P4].

Marriage was a concern for most of the participants. They believed there are some barriers such as physical impairment and financial issues and having a job. One of them said: *“I did not think of marriage at all, but now I think, but it’s an absurd thought, I still think that nobody has yet come to understand and cope with a person who has a lot of problems and so many differences because it needs profound thought”* [P5]. Regarding concerns about childbearing, the participant who married last year said: *“each woman wants to become a mother one day. Feels being mother inside. My problems for childbearing are gaining weight and it worries all family members especially my husband.*



*I don't Know I can climb the stairs or get up when I gain weight for pregnancy?" [P2]. For this reason, this participant stated in his interview that: He has a kind heart. Not for me. very big heart. He is a sincere and honest man. When I look at him at the top above. I think he is a pity one that has married me. Maybe he cannot become a father at all."*

Her husband is satisfied for marrying in spite of her problems. He said: *"I'm satisfied with one hundred percent. I'm so happy to have her. I am glad that could not make a difference between the sick and healthy people. It's not a disease, of course, is a problem that has come about. This thought that people has about these cases in the community should be removed. This culture must be make that such small issues should not be greatly enhanced. And for the sake of such a small topic, we should not leave them alone in community and quarantined them."* [P6].

### Uncertain prospect

The participants had no positive view of their future. They considered that as an unclear subject they are not able to plan for the future. One of them said: *"The future is ambiguous for me. I just want to be successful in every field I can ... at least be proud of myself..."* [P5]. One of them believed that he cannot make a plan because the situation is not very clear here. *"If you look realistically, it's very difficult to plan for the future .....you cannot make a plan.the situation is not very clear in the country. I always worried about the future because I don't know what's going to happen."*

## DISCUSSION

This phenomenology study gives valuable information about the day-to-day world of those who live with SMA and discloses the emotional dimension of physical dysfunction such as feel of shame, humiliation, and helplessness. Participants' experiences disclosed that SMA has profound impact on quality of life and makes them dependent on other members of family for meeting their needs. The study of Ho HM *et al.* (2016) showed that participants with SMA type II and type III bravely faced all the challenges and lived an autonomous life.<sup>[11]</sup> The experience of chronic illness affects all aspects of a person's life such as mental and social. They struggle with their pains and suffering during their lifetime.<sup>[14]</sup>

For the patients with disability, the impairments of body function can cause restrictions in their performance, tasks, and social roles which may then lead to social exclusion.<sup>[15]</sup> For society, disabled people is not only the burden on health and social services, but also they lose the opportunity of utilizing human resources. Another issue for many disabled people is the additional time and activities for maintaining an independent life such as washing, dressing, and cleaning, whereas nondisabled people take them for granted.<sup>[15]</sup>

Dependency is another issue that affects the life of SMA patients. They have problems for finding a suitable job because they have physical impairment and yet they were increasingly viewed as less productive in the workplace than their nondisabled counterparts.<sup>[15]</sup> If disabled people have a job, they can earn money and overcome the feeling of uselessness and their self-esteem will be improved. Araghian *et al.* showed that families with SMA face financial concerns.<sup>[6]</sup> If patients have income, they can feel better in family because it lessens the feel of dependency. In this study, participants declared that having a job reduces the burden for marriage and making a family.

Participants also stated "Shame of looking by others," that is it is widely acknowledged that the greatest impediment to the lives of young people with disabilities is prejudice, social isolation, and discrimination.<sup>[16]</sup> While all individuals with disability may be affected by this lifelong cycle of stigma and prejudice, females are at an increased risk.<sup>[17]</sup> In societies where girls are valued less than boys, the investment in education, health care, or job training that families are willing to make in disabled girls is often substantially less than that for disabled boys.<sup>[18]</sup> In this study, patients preferred to stay home and not accompany their friends or continue education for not be seen. These young people are less likely to be included in whatever services and programs that exist. They need a safe and supportive environment, education, health services, and access to sport and recreation. They also need to develop skills that will serve them well in the community and the workplace.

For some, specific physical or intellectual impairments may further limit their job options. For most, social prejudice makes employers hesitant to hire them.<sup>[19]</sup> Rates of unemployment among adults with disability vary from country to country, but, on an average, tend to be about 40%–60% higher than for the nondisabled population.<sup>[20]</sup>

In progressively degenerative diseases, time may also be disrupted in another way. Given the nature of such disorders, the future assumes an inherently problematic quality. Since the severity of the progress of the disease is not clear enough, it is difficult for patients to have long-term goals and plan for the future. Moreover, the future is not only problematic, but also overtly threatening. This change in the relation to the future contributes to personal meanings, in particular, one's sense of what is possible in one's life.<sup>[21]</sup>

In HO *et al.*'s study, SMA patients made self-adjustments in their lifestyles and exerted themselves to positively cope with hardships and maintain dignity and self-control,<sup>[22]</sup> contrary to the present study (in Iran) where people

involve with disappointment and limitation. The reason of difference can be the dominant values, social, cultural, economical, and welfare services of these two countries.

## CONCLUSIONS

This study was an attempt to look at the experience of patients suffering from SMA. We found that SMA patients describe their condition as severely disabling and dependent on others. SMA patients were very worried about their future, complain of God, and they know themselves as a victim of fate. Meanwhile, they considered their illness to be a divine destiny, trying to accept their condition, and provide psychological relief by comparing their conditions with patients in a much worse situation. They believed that vague future is waiting for them and they are not able to make plan. The study findings can be used as a resource for nurses in caring for SMA patients. Because of physical impairments, patients with SMA are dependent on family members to meet their needs, which creates a great burden for the family. Nurses should play crucial roles as specialists, planners, managers, and consultants to help patients and their family members.

## Conflicts of interest

There are no conflicts of interest.

## Authors' contribution

All authors contributed to this research.

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