

REVIEW

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Psychological burden in spinal muscular atrophy patients and their families: a systematic review

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Abstract

Background: Spinal muscular atrophy (SMA) is an autosomal-recessive disorder that manifests in paralysis and skeletal muscle weaknesses. This neuromuscular problem is caused by degeneration of the spinal cord anterior horn cells.

Main body of the abstract: SMA leads to a decreased motoric function that affects patients' daily activity and eventually induces the psychological burden. Given the increasing psychological burden of the patients and their families, this review summarizes the studies assessing the psychological burden of SMA patients and their families. We aimed to highlight the leading causes of their psychological burden. We systematically searched literature from the period 2011 to 2020 in two electronic databases by using the keywords "Spinal Muscular Atrophy", "Psychology", "Caregivers", and "Burden", then filtered it with the eligibility criteria and screened the abstract of the studies. By searching, screening, and filtering the studies, we included seven studies in this review. This review showed an increasing psychological burden in SMA patients, moreover for the parents. The dependency mainly causes the psychological burden since they eagerly struggle to achieve an everyday life.

Conclusion: Health professionals' understanding capability regarding the mental condition of patients and their families is a potential aspect that can create better care. This review is encouraging for researchers and clinicians to have an enormous understanding of SMA.

Keywords: Caregiver, Family, Psychological burden, Spinal muscular atrophy

Introduction

Spinal Muscular Atrophy (SMA) is an autosomal-recessive disorder induced by the homozygous deletion in the survival motor neuron-1 (SMN 1) gene on chromosome 5q13 [1, 2]. This problem leads to degeneration of the spinal cord anterior horn cells. This condition causes muscle weakness, paralysis, and atrophy [3]. Although the incidence is rare, 1 in 6000 to 11,000 live births [4–6], SMA is the most common genetic disease causing death in infants. The manifestations can occur in months to years,

depending on the type of SMA [7]. SMA in infants may lead to a decreased life expectancy that related to the low survival rate [1, 7].

Based on their phenotypes, SMA is divided into four types. SMA type-1, Werdnig–Hoffmann disease, is characterized by the symptoms that usually appear in infants under six months. They cannot sit or maintain their head straight. Symptoms in SMA type-2 appear between the ages of 7–18 months, while in SMA type-3 or Kugelberg–Wielander syndrome, a child can mostly survive until adulthood and walk [8]. SMA type-4 is an adult-onset type [7].

The mortality of SMA type-1 and type-2 is commonly due to respiratory failure because cell degeneration affects respiratory muscles. It leads to mucus accumulation and easily induced infection. Moreover, it

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is simultaneously accompanied by a decreased ability to cough [5]. SMA type-3 has an increased risk of falling since they lost their balance. The patient must struggle to deal with a gradually decreasing motoric function. Thus, it will limit their ability to do daily activities [6].

Therapy in SMA focuses on increasing the patient's life expectancy and quality of life [10]. Patients tend to maintain their self-confidence and endure all of the difficulties during fulfilling daily needs. However, due to decreasing of physical abilities and motoric functions, patients often experience some periods of sadness, anxiety, frustration, and loneliness [11].

On the other hand, the family also experiences physical, emotional, and psychosocial problems [12]. After they were diagnosed, the family must try to deal and be ready if their children's health got worse than before or when they need to make a complex decision related to medication or clinical interventions of their children. They need to give long-life supportive care. Thus, these things will make the family physically and mentally exhausted [13].

Since SMA treatment needs multidisciplinary experts, physicians need to know details from the patient's or family's aspect of a worsening SMA [14]. This review aimed to identify the events experienced by families and patients with spinal muscular atrophy and their psychological factors to provide information for families, patients, health workers, and researchers.

Materials and methods

This review aimed to conclude and echo the findings from the psychological aspect using a scoping review. In practice, this review was directed utilizing a methodological framework compiled by Arksey and O'Malley [15].

The populations we used were spinal muscular atrophy patients, defined as individuals with SMA disease; families defined as parents, siblings, or relatives; and caregivers defined as people who took care of the patient. The outcomes we used were a psychological burden, which is defined as emotions such as concern, anxiety, uneasiness, and depression. Anxiety can be interpreted as uneasy or anxious feeling, and afraid; depression, which is defined as gloom; and frustration, which is defined as feeling disappointed due to failure to do something.

Identification and selection of studies

The literature was identified from two electronic literature databases, including that were Pubmed and CINAHL. Studies from the period 2011 to 2020 were included in the keywords or Medical Subject Headings (MeSH) of "spinal muscular atrophy" or "spinal muscular atrophy in childhood", "psychological burden" or "psychological dependence", and the keyword "sadness",

"depression", "psychosocial", "palliative care", "quality of life", "experience", "family", "caregivers", and "siblings".

The studies obtained from the search results were sorted into two levels of screening. At the first level of screening, the studies were collected based on the requirements of the inclusion criteria. It was declared eligible based on screening the title and abstract. The study obtained must be qualitative or quantitative research published in 2011 up to 2020, and SMA patients were the subject. A viable study could not be included in this review if it was an opinion, a recommendation in management, a letter to the editor, a review, or not available in English or Indonesian.

Through an overall text review, studies were screened using the same inclusion and exclusion criteria at the second screening level. The cumulative articles included in this review at each level of the filtering process are shown below (Fig. 1).

Charting the data

The authors determine a summary of the studies that are included in this review. The summarized literature will be presented in the authors' name, study location, the population, objectives, and the findings related to psychological burden. These studies can be seen in Table 1.

Compiling, summarizing, and reporting the results

Summaries and conclusions drawn from each literature relating to the patients' and parents' experiences and impressions of SMA are summarized in a narrative form. The relationship between these findings was described in the context of the objectives of this review.

Consultation

After formulating and compiling the summary of this review, the authors conducted consultations and discussions about the findings and conclusions to find agreements and summarize all studies.

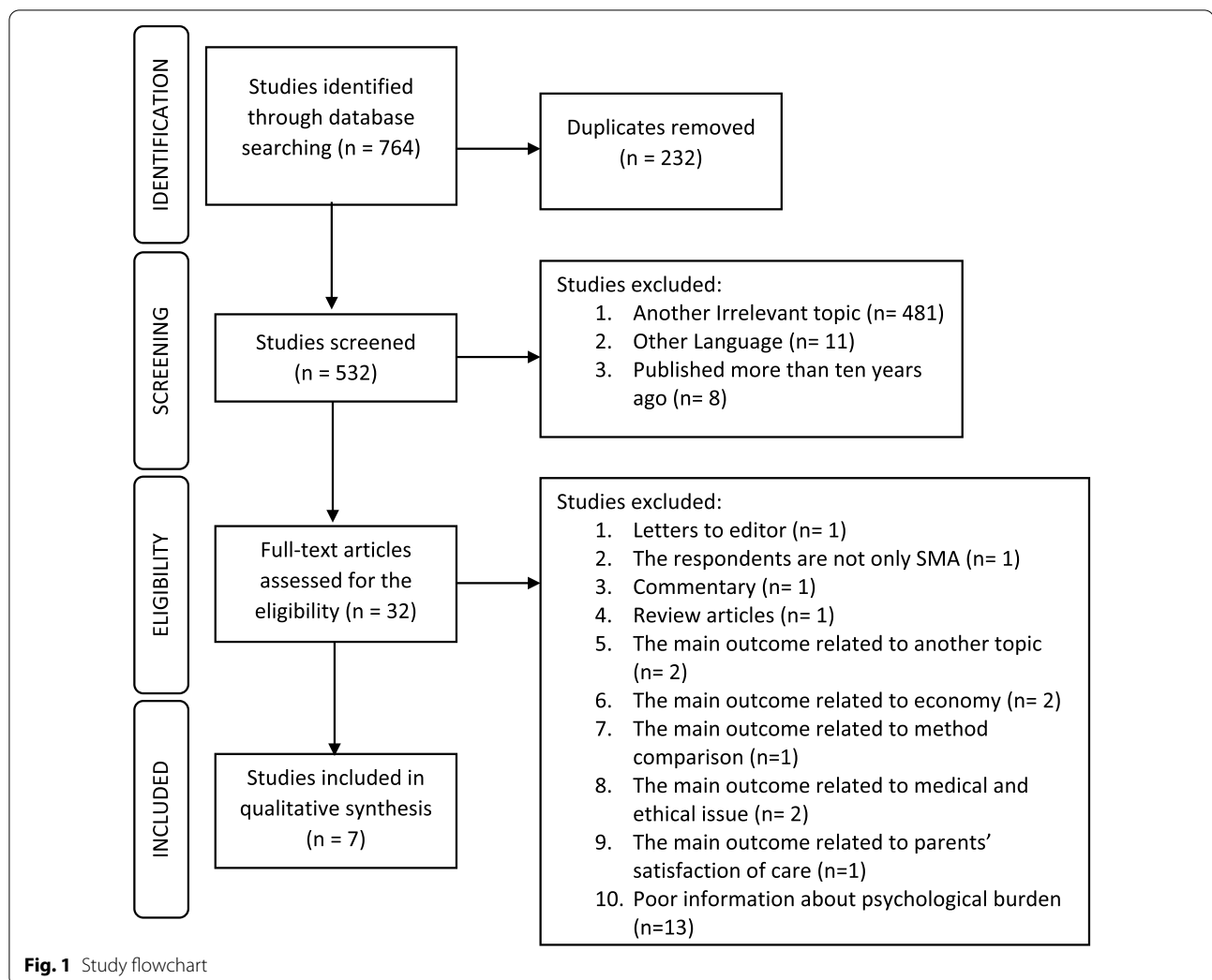
Results and discussion

Psychological burden of SMA patient

In terms of their illness, SMA patients have different conditions depending on the type of disease. The psychological burden in patients who experience symptoms since childhood will differ from those who manifest symptoms in older age.

Dependency on the caregiver

In a study conducted in Taiwan, one of the SMA type-2 patients said that parents always help them even in the most basic activities such as urinating. Parents support the patients by lifting them from a bed into a wheelchair and taking them to the toilet. After that, the



parents take the patient to the bed. They needed to be helped in their daily activities. The patients' limitations on basic physical abilities cause a burden to the family due to their dependency [9].

The study of five patients with SMA type-3 in Iran showed that they were worried about their aging parents' condition and did not want to interfere with their siblings' personal lives [18].

These findings illustrate that patients find it difficult to live without other people's help in their daily lives. Patients also realize that parents as caregivers will feel tired over time. Patients show their concern for the various things that are considered burdensome to their parents. However, patients also realize that they cannot be independent. It causes loss of self-confidence and dependency.

Being different

A study of 5 patients with SMA type-3 conducted in Iran explained that patients often feel embarrassed when others are noticed. In that case, the patient always tries to ignore it. Another patient said that patients avoided the opportunity to write on the blackboard to hide their illness while at school. However, patients have already anticipated that opportunity. Patients also feel burdened when their classmates find out about the disease [18].

In the study, patients also explained that their health conditions prevented them from taking many opportunities to attend school, pursue hobbies, or gather with peers. Patients also felt sad and guilty for making much trouble to the family and regretted being born. During adolescence, the patient sometimes feels very miserable. The patient compares himself to his siblings and

Table 1 Studies included in the review

No.	Author	Study location	Population studied	Study objective	Findings
1	Ying Qian et al. [6]	USA	21 individuals with SMA (age: 8–26 years; mean age of diagnosis: 2.05 years; type-1: 1, type-2: 8, type-3: 12); 64 parents of individuals with SMA (type-1: 12, type-2: 29, type-3: 22) 11 Clinicians	To investigate: 1) factors influencing how families arrive at a diagnosis, as described by parents and clinicians; 2) parents' perspectives on newborn screening; and 3) the impact of SMA on the lives of people with SMA and their parents	Parent's experience: they feel sad to see the decreased function of their child compared to other normal children
2	Hsin-Mei Ho et al. [9]	Southern Taiwan	9 patients with SMA (age: 25–54 years; age of onset: 1–14 years; type-2: 2, type-3: 7)	To explore lived experiences of patients afflicted with SMA	Patient's experience: they feel sad and stressed due to their inability to be independent
3	Emily J Higgs et al. [10]	Australia	13 individual parents representing 7 families of SMA type-1 patients (age at diagnosis: < 1–5 years; age at death: < 3–12 years)	To examine parents' perspectives of having a child with SMA type 1, from diagnosis until loss of their children, to inform clinical practice by identifying the most meaningful aspects to parents to develop a supportive strategy	Family's experience: they feel angry, frustrated, and depressed due to the disease's diagnosis and familial history aspect. They blame themselves
4	Bao-Huan Yang et al. [14]	Taiwan	19 parents of children with SMA (age: 9–12 years; the age of onset: 4 month–3 years)	To explore the parents' anticipatory loss of school-age children with type-1 or type-2 SMA	Parent's experience: they feel sad for the uncertainty of their child's future and death
5	Sally Lawton et al. [16]	Australia	6 interviewed and 28 surveyed relatives of SMA patients (type-1: 7, type-2: 8, type-3: 4)	To investigate family members' perspectives on the journey to receiving a diagnosis. To explore their views on the potential of earlier diagnosis	Parents' experience: they feel devastated and denied the uncertain diagnosis and the disease's outcome. They feel frustrated about the future of their child
6	Ali Asghar Jesmi et al. [18]	Bojnord, North of Khorasan province, Iran	5 patients' SMA type III and one patient's wife (age: 26–32 years)	To investigate the lived experiences of SMA patients	Patient's experience: they feel shame and blame the disease due to their inability to do a regular thing. They feel helpless when thinking about their future
7	Michele A Farrar et al. [17]	Australia	7 primary carers of 8 children with SMA (mean age: 6.4; type-2: 5, type-3: 3)	To gain insights into the effect caring for a child with SMA has on financial (both direct and indirect), opportunity, and psychosocial costs associated with SMA care from a primary carer perspective within the Australian health system	Family's experience: they feel helpless as the SMA forces them to continuously worry and limit them to have recreation due to the time-consuming taking care of them

* USA United States of America, SMA spinal muscular atrophy

complains about the conditions considered unfair to God. In several times, he tries to commit suicide [18].

This study shows the reality felt by SMA patients who have significant limitations that make them different from other people. Patients often experience disappointment because their physical abilities cannot support their desire to take some opportunities they have hoped for. One of the differences they received is a form of injustice towards them that caused a deep sense of sadness.

Uncertainty and hope of the future

A study conducted on five patients with SMA type-3 in Iran showed that there were feelings of helplessness. They felt suffering when talking about the future. Patients felt that they had little hope for their future. They presumed it to be gloomy and ambiguous. Patients feel unable to make a plan for their future. Another study of parents of children with SMA type-1 and type-2 conducted in Taiwan explained that children feel afraid and helpless about death [14].

This study explains that patients are concerned about their condition and survival rate that cannot be ascertained. Their physical condition will decline in the future, reducing their mobility and potential development or improvement.

Psychological burden of SMA patients' family

Families are the main person who supports and takes care of the SMA patients. The more their children are dependent, the more they want to help their children endlessly up to their limit. Witnessing their children grow, develop, interact with the environment, and have some changes in their physical abilities, parents run to the point when it reaches the highest emotional stress level due to the inability to provide the best treatment for their child. Somehow, parents often feel sad, angry, and frustrated while accepting the reality of this condition.

Being aware of the diagnosis and the declining condition of the child

A study conducted in Australia on 13 parents of children with SMA type-1 stated that they were already aware of hypotonia symptoms, lack of head control, and delayed motor development from birth until three months old. This condition creates confusion for parents and many health professionals who are not aware of SMA disease. Children were diagnosed with SMA at the range less than one year old to 5 years old through the collaboration of neurologists, pediatricians, and counselors [10].

When parents receive an explanation from health professionals about the symptoms and clinical manifestations of their child's illness, they tend to refuse to believe. They were shocked and saddened by the child's inevitable

death [10]. In a study conducted in America, parents feel sadness. They had stress watching their child experiencing the disease and worsening [6].

A study in Taiwan also illustrates that patients and their families feel very sad and stressed after knowing that this illness cannot be cured [9]. Moreover, a study conducted in Australia added that parents tend to refuse the diagnosis because they are uncertain about the conditions that occur to their children. It causes anxiety, worry, and stress [16].

To obtain a diagnosis of SMA, patients or parents commonly visit many different doctors to confirm the diagnosis but do not get the desired results. These findings indicate that parents experience a period of confusion about their child's condition for a long time and eventually lose hope, especially when they receive an explanation about SMA. It causes severe and prolonged mental stress.

Being aware of the future of the child

A study conducted on 19 parents of children with SMA type-1 and type-2 school-age children showed that parents often feel sad about their child's uncertain future. Parents realize that when their child is getting older, they are closer to death. They felt sadness and a sense of helplessness coupled with anxiety because they might lose their children at any time [14]. Another study conducted in Australia shows that most parents are always hopeful to think about their child's health in the future, while some parents feel stressed, worried, and frustrated because they are pessimistic about their child's future [16].

Information regarding the children's life expectancy makes the parents sad because they cannot prevent this disease from worsening while the time is running out. These findings illustrate the psychological burdens of anxiety, sadness, frustration, and hopelessness of parents. Mostly, parents hope to create a better future for their children.

Time consuming

A study conducted on seven caregivers of patients in Australia found that families who focus on sick children may fatigue and waste too much time. As a caregiver, the family should be around their children all day and night without ever leaving their side for a long time. The same research also states that the parenting role seems to be transformed into caregiving [17].

The condition of the highly dependent patient on a wheelchair causes a loss of spontaneity in daily activities. Somehow, this disease indirectly affects the marriage relationship's dynamics because parents play a significant role in caring for their children. It creates high tension

among parents. Parents feel they do not have enough free time to tighten their relationships and hang out with friends. A study conducted in the United States claimed that the parents' conditions encouraged them to accept that they would lose time and opportunities to do activities with friends [6].

Taking care of SMA patients forces the caregivers to devote more time. These findings illustrate that parents feel tired over a long period and do not get enough time for recreation to reduce stress or calm, motivate and strengthen their mental strength.

Being aware of the risk of having another child

The Australian study demonstrated a concern from one of the parent participants about her plans to conceive again. Parents feel a loss of joy and reproductive freedom when discussing plans to get pregnant again after losing their child to SMA. The same research also shows parents' sadness who blame themselves because they feel it was their fault. They think that this disease results from a genetic problem passed down by them [10]. The study illustrates parents' heavy burden in planning a new pregnancy after knowing the risks of their child who may develop the same condition.

Conclusions

In the struggle to live as SMA patients, they will have various limitations that cause them not to carry out social activities and do their hobbies. The family as caregiver is responsible for helping them to cover up the limitations. Emotionally and psychologically, the patient receives pressure from his limitations, which significantly impact himself, the people surrounding him, and his hopes for the future. Families, especially parents, also receive pressure from their helplessness in creating an everyday life for patients.

This review concludes that the psychological burden happening in SMA patients is generally developed by the patient's dependency on the caregiver, being different from ordinary people, and the uncertainty of their hope for the future. On the other hand, the psychological burden that happens in SMA patient's families developed due to the process of understanding their child condition and diagnosis, the possibility of child's future, the risk of the same disease when having another child, and the plenty of time wasted due to the child's condition.

Health professionals' understanding capability regarding the mental condition of patients and their families is a potential aspect that can create better care. In SMA patients' care, intervention is required to slow down the disease, prevent complications or a worsening stage, and maintain the family and the patient's mental and psychological health. In the following study, we have to find

the most effective method to support SMA patient's and their families' mental health. However, this review summarizes a small number of studies conducted in several countries around the world. Therefore, it only points out a common condition in patients and their families. It cannot be used as a standard to represent all SMA cases worldwide. Another limitation is that no study discusses the psychological burden between patients and uses psychometry scales to measure depression and anxiety.

Abbreviations

SMA: Spinal muscular atrophy; SMN: Survival motor neuron.

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Author contributions

WD and MM collected articles and references. WD, MM, DM, and LC discussed and organized the content in the manuscript. All authors read and approved the final manuscript.

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Availability of data and materials

The data sets used and/or analyzed in the current study are available from the corresponding author upon reasonable request.

Declarations

Ethics approval and consent to participate

Ethical approval is not applicable in this review.

Consent for publication

Consent for publication is not applicable in this review.

Competing interests

The authors declare that there is no conflict of interest.

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