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2016

### **document version**

Publisher's PDF, also known as Version of record

[Link to publication in VU Research Portal](#)

### **citation for published version (APA)**

Bakker, M. (2016). *Moving together towards participation: Insights into illness and care experiences of people with neuromuscular diseases*.

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**Illness experiences in neuromuscular  
diseases: a systematic review**

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UNDER SUBMISSION AT DISABILITY AND REHABILITATION

# ABSTRACT

**Purpose:** Most studies on neuromuscular diseases (NMDs) concentrate on biomedical issues. However, research on illness experiences is increasing. The purpose of this systematic review is to provide an overview of the existing knowledge about people's experiences of living with NMDs. This can help professionals to better understand the care needs of people with NMDs.

**Method:** This study focused on three NMDs: Post-Polio Syndrome (PPS), Facioscapulohumeral muscular dystrophy (FSHD) and Amyotrophic Lateral Sclerosis (ALS). These NMDs were selected because of their diversity in progression, age of onset, and symptoms. A literature search was performed in the bibliographic databases PubMed, Embase.com, PsycINFO (via EBSCO), CINAHL (via EBSCO) and the Cochrane Library (via Wiley). We searched for subjective, daily experiences. The included studies were analysed thematically.

**Results:** Twenty studies met the inclusion criteria. Themes that emerged were: experience with diagnosis; experience with symptoms; quality of life; use of devices; coping; and, for ALS, decisions concerning end-of-life. Most studies focussed on the experience of the physical and mental state of people. Less attention was paid to the social consequences of being disabled. Social consequences include i.e. loss of independence.

**Conclusion:** In literature on illness experiences with NMDs, there seems to be a lack of attention on the social consequences of being disabled. Addressing these personal social aspects in rehabilitation could improve participation in society.

**Keywords:** patient perspective, post-polio syndrome, amyotrophic lateral sclerosis, facioscapulohumeral muscular dystrophy, quality of life, literature study

## INTRODUCTION

Neuromuscular diseases (NMDs) are rare, chronic diseases that can have a large impact on many aspects of life. In NMDs, muscle function is impaired and declines over time. There are many different NMDs, all with different progression rates, age of onset and rate of pathophysiology, but all are characterised by increasing muscle weaknesses. This can result in the loss of mobility up to the loss of independent walking ability and total dependence in daily living activities (1; 2). In this perspective, a NMD is, besides a chronic disease, a progressive disability that affects different aspects of peoples' lives.

Studies have focused on aspects of NMD related to physical disability (1), treatments or interventions (3-6) and quantitatively measured quality of life (QoL) (7-9). These studies are all characterised by quantitative research methods.

Although the majority of research about NMDs concentrates on quantitative measurements, the amount of qualitative research regarding illness perceptions and illness experiences is increasing. This can be explained by the increase of rehabilitation articles, that focus on participation in society rather than biological aspects, QoL or treatments. For rehabilitation the illness perceptions and illness experiences are eminently important, as these can serve as a basis for a deeper understanding of aspects of patients' lives and can assist in problem solving (10).

A way to study illness perceptions and experiences is by means of phenomenological research (7; 11). Phenomenological research focuses on the 'lived' experiences of persons and takes the insiders perspective and context of the respondent into account (11; 12). It aims to describe the experience of everyday life and its meaning. Phenomenological descriptions "are possible only by turning from things to their meaning, from what is to the nature of what is" (13, p. 191). As the context influences the meaning of the experiences, context is deliberately taken into account (14; 15).

Despite the increase of phenomenological studies on illness experiences of people with NMDs, an overview of these experiences is currently lacking. Such an overview is relevant for the rehabilitation practice, as it can help professionals to better understand the needs of people with NMDs and improve the rehabilitation care based on these needs. Therefore, a systematic review about the illness experiences of people with NMDs was executed. The purpose of this article is to gain an interpretive understanding of the subjective impact of NMDs on the lives of patients, as revealed by a systematic review and synthesis of literature on illness experiences in patients with NMDs. Additionally, such an overview and analysis can help to identify new research topics and approaches.

# METHODS

## Design

The PRISMA statement was followed for this systematic literature search (16). The main purpose was to provide an answer to the question of 'what is known about patients' experiences with living with NMD?'. MB and RO developed the review protocol and performed the literature searches. MB and KS critically discussed the review process and the results. FN and TA were involved in the analysis and interpretation of the results.

## Search and selection process

This systematic review falls in the scope of the larger FACTS-2-NMD program, in which rehabilitation interventions in 3 NMDs were studied (17-19). This larger program selected three different types of NMDs, based on their variety in age of onset, rate of progression, and pathophysiology, and, therefore, are assumed to represent and give insight into a range of experiences with different types of NMDs. The three NMDs are Amyotrophic Lateral Sclerosis (ALS), Facioscapulohumeral Muscular Dystrophy (FSHD), and Post-Polio Syndrome (PPS) (see box 1 for more information about the diseases).

### Box 1: description of diseases

ALS is a disease characterised by progressive degeneration of motor neurons in the brain and spinal cord resulting in muscle weakness. The median age of onset of ALS is in the mid-fifties. Initial symptoms are weakness of limbs or weakness in the bulbar region causing speech abnormalities, swallowing difficulties and facial weakness. Due to progressive muscle weakness, the patient eventually becomes paralyzed. Approximately 50% of patients die within three years of onset, usually due to respiratory failure (60). Globally, the median prevalence of ALS is 4.48 per 100,000 (61).

PPS also acts on the motor neurons, but progression is very slow. PPS symptoms usually start thirty to forty years after acute polio. After recovering from the acute stage, patients often were encouraged to push themselves to limit the impact of impairment upon their lives. Many individuals with PPS are overachievers, who, despite their lasting and increasing physical disabilities, deny new symptoms and suffer from overuse. Approximately 25% to 50% of polio survivors will be affected by PPS. In 2012, the US alone counted 1.6 million polio survivors (62).

FSHD is an inherited neuromuscular disease with the onset of symptoms occurring between six and 20 years of age. It is a slow progressing muscular dystrophy that mainly affects the muscles of the face, shoulder blades and upper arms. With time and depending on how severely an individual is affected, the leg muscles may become affected as well. FSHD is one of the most prevalent muscular dystrophies, with a prevalence of 1: 15.000–1: 20.000 (63).

A literature search was performed by RO and MB in the bibliographic databases PubMed, Embase.com, PsycINFO (via EBSCO), CINAHL (via EBSCO) and the Cochrane Library (via Wiley) from inception to the 2<sup>nd</sup> of June 2015. We searched for the subjective/daily experience in three chronic muscular diseases (PPS, FSHD and ALS). Search terms

included controlled terms (e.g. MeSH in PubMed and Emtree in Embase) as well as free text terms. We used free text terms only in The Cochrane Library. Search terms expressing daily/subjective experience were used in 'AND' combination with search terms comprising PPS (postpoliomyelitis syndrome), FSHD (facioscapulohumeral dystrophy) or ALS (Amyotrophic Lateral Sclerosis). The full search strategies for all the databases can be found in the appendix.

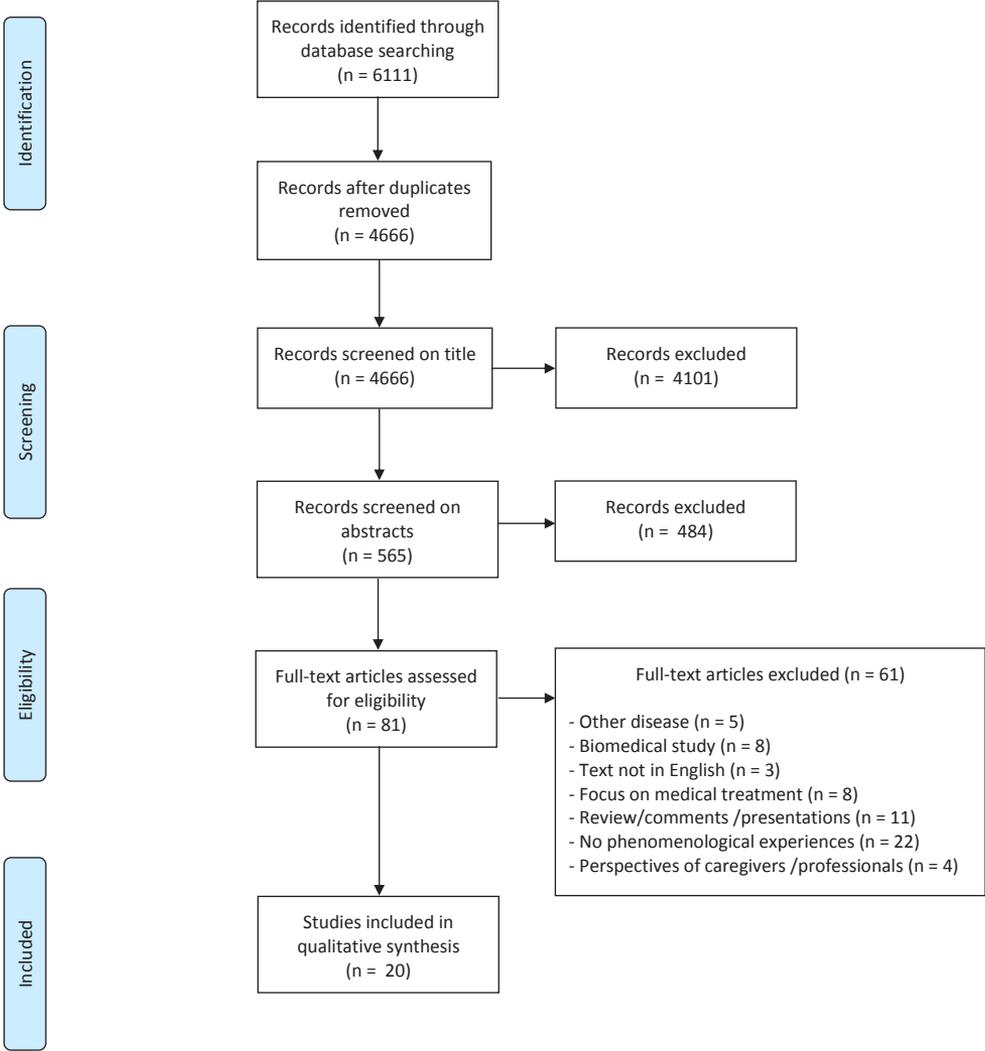


Figure 1: Flow diagram

The retrieved articles were transmitted into Reference Manager. From there, a copy of the titles, abstracts and keywords of the retrieved articles were transmitted into a PDF file. To improve objectivity, author names and journal titles were blinded. Appropriateness of inclusion was judged in different rounds; in the first round, selection was based on the titles. In the second round, titles and abstracts were read and in the third round the full texts of the articles were read. Inclusion criteria were that articles reported on the lived experience of PPS, FSHD, or ALS from the patients' perspectives. The studies had to use a research design that allowed an in-depth exploration of personal experience. Quantitative and mixed methods studies were included in the search but did not meet this criterion. We included articles that reported on the experience of both people with NMDs and their caregivers, but articles that reported solely on the perspective of caregivers were excluded. Theoretical articles were excluded, as well as essays, editorials, thesis and book reviews.

Common reasons for exclusion were a focus on effectiveness of interventions, a lack of phenomenological understanding of the meaning of experiences, or a focus exclusively on caregivers, see figure 1. Following the review of title, keywords, and abstract, 20 articles were retrieved and reviewed.

### **Synthesis of data**

Because of the phenomenological focus of the study designs, a statistical synthesis was not appropriate. Therefore, a narrative synthesis was undertaken (20). All included data were entered into the qualitative software MAXqda and analysed by theme. MB conducted a preliminary analysis of each selected study, critically appraised each study and explored similarities and differences between the studies. MB,KS, TA and FN then synthesised and interpreted the evidence as it related to the purpose and aims of the review.

No attempt was made to reanalyse primary data presented as quotations, as these were inevitably sparse and selective in relation to the totality of the data in any study.

## **FINDINGS**

In total, using the above search terms, 20 studies were included in the analysis. Fifteen studies focused on ALS, five on PPS, and none of the articles focused on FSHD. All articles were published in peer-reviewed journals. Different types of articles were found. Most studies used qualitative interviews and data analysis. One article was based on a patient's story (21). As all selected studies used phenomenological experiences as data, they are equally valuable for this review.

Based on the analysis of the articles, we have categorised the studies under the following headings: the diagnosis, experience of symptoms, use of devices, Quality of Life (QoL), coping strategies and end of life. Reported illness experiences differ between the diseases and will, therefore, be described separately. Studies are also summarised in Table 1.

**Table 1: Included articles**

Author and Year	Illness	Method	Data Analysis	Aim and results
Atwal et al., 2014	PPS	Focus groups	Thematic analysis	<p><i>Aim:</i> to gain an in-depth understanding of the meaning of quality of life for polio survivors and to determine the type of strategies that are used by people with PPS and the support that they consider as important to facilitate participation in everyday life activities that have an impact of their quality of life.</p> <p><i>Results:</i> The research identified resolvable factors that influence quality of life namely inaccessible environments, attitudes of health0care professionals and societal attitudes.</p>
Bolmsjö, 2001	ALS	Semi-structured interviews	Hermeneutic method	<p><i>Aim:</i> to study how patients with ALS communicate when existential issues are touched on, and what kind of problems the patients experience dealing with such questions.</p> <p><i>Results:</i> patients experience a number of problems, particularly in connection with physical inability and when the need to confide in someone is not particularly strong. Critical to the value of life is to be respected as a person. This study shows that existential issues are of great importance to the patients.</p>
Brown, 2007	ALS	Narrative interviews	Content analysis	<p><i>Aim:</i> to explore patient's experience and how they talk about living and coping with ALS.</p> <p><i>Results:</i> four types of storylines were identified: fracturing, preserving, enduring, and sustaining one's storyline.</p>
Charles, 1985	ALS	Personal Story	Experiential knowledge	<p><i>Aim:</i> to give a personal portrayal on the physical and emotional efforts that go into maintaining a "normal" way of life.</p> <p><i>Results:</i> an explanation of the issues that arise in the decision making process of respiratory support. Issues such as QoL, financial resources and a supportive family are described.</p>
Cobb et al., 1986	ALS	Extensive interviews and a number of standardised tests	Content analysis	<p><i>Aim:</i> to pay attention to the way that patients and families deal with the effect of ALS on their lives and to systematically describe the course of the illness from the patients' perspective.</p> <p><i>Results:</i> this research illustrates the importance of a link between professional and popular health care sectors since so much of the care of persons with ALS is given at home.</p>

Foley et al., 2007	ALS	Semi-structured interviews	Colaizzi's seven-step method for qualitative interview analysis	<p><i>Aim:</i> to explore the self-referent meaning of QoL as perceived by individual ALS patients, and to explore how their experience of health care affected their subjective well-being.</p> <p><i>Results:</i> physical decline is not experienced as the main problem as it does not necessarily result in a decreased QoL.</p>
Foley et al., 2014	ALS	In-depth interviews	Grounded theory	<p><i>Aim:</i> to identify process that underpin how and why people with ALS engage in health care services.</p> <p><i>Results:</i> The study gave insight into how people with ALS adapt to loss. People with ALS live with insurmountable loss and never regain what they have already lost. Loss for people with ALS is multidimensional and includes loss of control. The experience of loss of control prompts people with ALS to search for control over health care services.</p>
Hansson et al., 1999	PPS	Semi-structured interviews	Inductive analysis	<p><i>Aim:</i> to describe the illness- related problems experienced by people with PPS in their daily lives and how they cope with these problems.</p> <p><i>Results:</i> it was found that the participants experienced many types of illness-related problems in their everyday lives. Largely, the participants have learned to live with the changes and feel that they have 'a good life in spite of everything'.</p>
Jönsson et al., 1999	PPS	Semi-structured interviews	Grounded Theory	<p><i>Aim:</i> to gain a deeper understanding of how persons with Polio manage their daily occupations in order to adapt to environmental demands.</p> <p><i>Results:</i> the strategies were interwoven, revealing different ways of reaching an adaptive balance in daily occupations for the present and the future.</p>
King et al., 2009	ALS	In-depth interviews, field notes, electronic correspondence, stories, prose, songs and photographs that are important to participants.	Grounded theory	<p><i>Aim:</i> to present a model that explicates the dimensions of change and adaptation as revealed by people who are diagnosed and live with ALS.</p> <p><i>Results:</i> the lives of people living with ALS revolve around the need to make decisions about how to live with the disease progression and their deteriorating abilities. Life decisions were negotiated by participants.</p>
Lemoignan et al., 2010	ALS	Qualitative phenomenological methods using semi-structured interviews	Content analysis	<p><i>Aim:</i> to better understand the experience of decision making about assisted ventilation for ALS patients.</p> <p><i>Results:</i> people with ALS and caregivers value autonomy in decision making about assisted ventilation. Their decision-making process is neither wholly rational nor self-interested, and includes factors that health professionals should anticipate and address.</p>
O'Brien et al., 2011	ALS	Narrative interviews	Grounded Theory	<p><i>Aim:</i> to gain a better understanding of the period from symptom onset to diagnosis from the perspective of those living with ALS.</p> <p><i>Results:</i> the diagnostic journey is not straightforward. People react differently to the diagnosis. However, the uncertainty is a burden on everyone.</p>

Oh et al., 2014	ALS	Observations and semi-structured interviews	Ethnographic approach	<p><i>Aim:</i> to explore the participants' illness experiences of patients with ALS in the sociocultural context of South Korea.</p> <p><i>Results:</i> The illness experiences were described in a 'journey of suffering' with three themes: 1) off the course, 2) drifting 3) a new boat. Participants experienced multidimensional suffering as the disease progressed.</p>
Ozanne et al., 2013	ALS	Semi-structured interviews	Content analysis	<p><i>Aim:</i> to illuminate how people with ALS create meaning despite the disease.</p> <p><i>Results:</i> The uncertain journey towards death was more frightened than death itself. Despite the incurable disease, people found meaning in life; through their families and friends, the act of giving and receiving help, the feeling of having a life of their own and accepting the present.</p>
Rosengren et al., 2015	ALS	Biographies	Narrative analysis	<p><i>Aim:</i> to describe patients experiences of living with ALS in the end-of-life situations.</p> <p><i>Results:</i> The themes 'suffering', 'meaningfulness', and 'experiences of a limited life' were identified as describing patients' understanding of living with ALS.</p>
Sundling et al., 2009	ALS	In-depth interviews	A qualitative content analysis method	<p><i>Aim:</i> to describe the experiences of patients with ALS as well as their caregivers of non-invasive positive-pressure ventilation (NPPV).</p> <p><i>Results:</i> improved sleep because of NPPV (resulting in a better QoL), but, at the same time, loss of independence. This was also difficult for the caregivers.</p>
Wenneberg et al., 2000	PPS	Semi-structured interviews	Content analysis	<p><i>Aim:</i> to further study the unique lifetime illness experience of persons with Post-Polio Syndrome.</p> <p><i>Results:</i> this study shows that Polio is a disease that not only touched persons deeply on a physical level but also on a psychosocial level, making them still vulnerable after 50 years of chronic illness.</p>
Widar et al., 1998	PPS	Interviews and questionnaire	Content analysis and quantitative analysis of questionnaire	<p><i>Aim:</i> to describe what pain means for persons with PPS.</p> <p><i>Results:</i> the study showed that people with PPS have tangible problems with pain.</p>
Young et al., 1994	ALS	Interviews (open and closed questions)	Mixed methods	<p><i>Aim:</i> to explore the issues regarding the use of mechanical ventilation from the patients' perspectives.</p> <p><i>Results:</i> the decision to continue life using mechanical ventilation or to forgo life-sustaining technology and allow death to occur is a daunting one. This study identified and explored some components of that decision-making process.</p>
Young et al., 1998	ALS	Interviews (open and closed questions)	Constant comparative approach	<p><i>Aim:</i> to explore the experiences contributing to a positive quality of life for people with ALS.</p> <p><i>Results:</i> themes that emerged were the use of cognitive reappraisal, reframing and intellectual stimulation as coping mechanisms, and the vital importance of interpersonal relationships.</p>

## Diagnosis

There is a difference between getting the diagnosis of ALS and PPS; ALS is a certain death warrant, whereas PPS does not influence the life expectancy. Therefore, the diagnosis has a very different meaning in the lives of the patients.

*ALS:* Several articles investigated getting an ALS diagnosis. One article specifically focused on the period from symptom onset to diagnosis (22) while other articles paid attention to the broader process of the diagnosis (23; 24).

A number of studies reported about the response of patients to the diagnosis. In general, the patients experienced fear, sadness, sorrow, loneliness and bewilderment (22; 23), sometimes resulting in denial (23). The fear concerned death itself, but also the process of deterioration, losing the ability to speak, and possibly becoming respirator-dependent in the later stage of the disease (25). Although traumatic, to some patients, the diagnosis also felt like a relief because they finally knew the origin of their symptoms (22). Some patients experienced a lack of information about the diagnosis and patient variability. Generic information about prognosis had unrealistic implications for those with atypical progression (22).

A delay in the diagnosis of ALS was not uncommon. Lack of urgency within primary care resulted in delayed referral for specialist investigations. Furthermore, delays occurred when patients were directed to specialists other than neurologists (22). Another reason for the delay was that patients did not recognise muscle weakening as a serious symptom of worsening health, but as a consequence of poor fitness (22).

Patients had mixed experiences with the communication of the diagnosis. Some patients experienced the process positively and stated that physicians were sensitive, gave good explanations, and were sympathetic; other patients experienced the communication as blunt and unsatisfactory—revealing poor communication skills and lack of consideration for the impact of the diagnosis on patients and relatives (22; 23).

Some studies reported that patients had difficulties telling the diagnosis to relatives (22). This related to the fact that patients would become dependent on their family; Western society's emphasis is on individualism, self-reliance and personal autonomy. Therefore, it is difficult for most people to imagine a life with disability and dependence on others (24).

*PPS:* Most articles paid attention to the diagnosis of the first polio onset. The experiences of this disease as a child had a great impact on patients. Required periods of isolation were especially traumatic to most patients (26). Little is written about the diagnosis of post-polio syndrome. Some patients experienced a lack of recognition of the disease by their physician (26).

In general, the studies show that the process of getting diagnosed is experienced as important by ALS as well as PPS patients. Recognition of the symptoms turns out to be an important part of the process and could more quickly lead to the definite diagnosis.

### **Experiences of symptoms**

The studies did not provide an overview of the symptoms experienced, but rather gave insight in how patients experienced certain symptoms.

*ALS:* Symptoms such as muscle weakness in the limbs and bulbar, and uncontrolled laughing or crying are well-known symptoms. Patients experienced these symptoms as troublesome. They often felt ashamed or humiliated by the loss of control over their body (27).

The relatively fast worsening of symptoms was often experienced as a stressful symptom by some patients (25; 27). Many patients described the fear of getting worse and, in the end, of the loss of control over the disease and body (27-30). The physical deterioration also led to the feeling of the loss of identity as people were unable to be who they wanted to be (considering e.g. social activities and work) (30). Also, one's social role changed when becoming more and more disabled. A South-Korean study described the importance of roles in i.e. the South-Korean culture. Being unable to maintain the role as head of the family was, at least in some cultures, very disturbing for the patient as well as their family (25).

Beside the loss of identity, patients were afraid of becoming a 'vegetable' and being completely dependent on family and health care (28). The hopelessness and uncertainty about the future (not knowing when the suffering will start and what it will look like), and the fact that the disease often progresses fast, without 'periods of normalization', makes ALS difficult to handle (28; 30-32). Once the symptoms get worse, the patients often felt ashamed of themselves and did not feel comfortable going out or meeting friends, which sometimes led to isolation (28).

*PPS:* For the majority of PPS patients, pain was a big problem. Pain experiences varied among patients and time, and cold weather influenced the severity of pain (33). Due to the pain, some patients experienced difficulty falling asleep while others found it hard to get up in the morning. Different causes of pain were described, ranging from a physical overload to a lack of recovery time (34).

Another well-known symptom was fatigue (26). Patients got frustrated that they had to plan their day carefully in advance and felt that they could not function normally due to the exhaustion (26).

Patients were often less mobile, which made them more dependent on others. This dependency was experienced as negative, and some felt like a burden to their relatives. Patients sometimes felt worthless, disappointed in themselves and angry (34).

Although the symptoms of ALS and PPS differ from each other, the negative feelings with regard to becoming dependent on family or health care are described by both patient groups. This consequence of the disease is, regardless of the type of disease, experienced as very difficult by patients, raising negative feelings such as anger, fear, disappointment and feeling a burden.

### **Use of devices**

Many patients with PPS use devices during the day like a wheelchair, cane, or other special devices in the home. None of the articles articulated how patients experienced the use of these devices. For ALS patients, experiences with a ventilator were deliberately investigated. Other devices such as a speech device or wheelchair were not described in the studies. Therefore, this section concentrates on the use of a ventilator by ALS patients, making a distinction between invasive and non-invasive ventilation (NIV).

*Non-invasive ventilation:* ALS patients may start to suffer from respiratory failure as the illness progresses. NIV is one way to palliate the symptoms of respiratory failure (35). Deciding whether and when to start with NIV was experienced by many as problematic. Patients had many considerations before starting their NIV: a fear of not being able to communicate, eat, or move (35), becoming dependent on a machine and losing autonomy (35). The decision to start NIV was sometimes complicated by the lack of information (36). In general, however, patients experienced a positive effect of the NIV, resulting in better sleep, less fatigue and being able to perform several activities such as at family events (36).

*Long-term mechanical ventilation (LTMV):* LTMV is given via a tracheotomy and is still the only intervention that prolongs the life of ALS patients by a matter of years (35). Deciding if and when to start with LTMV was explained as difficult for many. Considerations taken into account were fears like becoming dependent or not being able to communicate or move around (35; 37). Users of LTMV felt, however, and much more so than NIV, as though they were making a choice between life and death (35; 37). Some patients perceived the choice of LTMV as a burden (21). Patients considered to what extent they were a burden to their caregivers. Some felt like they were making their caregivers 'slaves' (37). An important reason for patients to start the LTMV was the fear of choking or running out of air and the feeling that they were not yet ready to die (35).

One article reported that the first phase of being on a ventilator can be a struggle for patients; they have to get used to a new kind of breathing, which can be difficult at first

(21). Once the decision for the LTMV was made, patients were generally positive about their lives. They saw the ventilator as their 'friend' (37) and felt that they could have a lot of valuable moments which they would not have had without the respirator (21). However, patients still hoped for a better treatment with a less cumbersome device (21).

Not many studies have looked into the experiences with the use of assistive devices. So far, only experiences with ventilation is described. These experiences show that it is difficult for patients to decide whether or not to start with ventilation, as it is difficult for them to estimate how the ventilation will limit their possibilities to participate in daily life.

### **Quality of life**

For PPS as well as for ALS, different studies investigated how the disease influenced the QoL of the patients. Although most studies on QoL were quantitative, some studies also reported phenomenological experiences of how the disease had affected the QoL of patients.

*ALS:* The experienced QoL was less straightforward than might be assumed. The QoL did not necessarily correlate to the physical ability of patients; QoL did not decrease as the disability increased (7). Instead, patients kept being grateful for the things they remained able to do and some became more grateful of their lives with their loved ones (32). Problems and negative thoughts were, however, also reported (21; 29). Patients felt they had nothing to give, and some started to feel useless or meaningless (25; 28; 29). Some were able to find a way to re-establish their self-worth; for example, by committing themselves to improve the facilities for the disabled or by having good conversations (27).

Informal support was experienced as important to patients' QoL. Some patients experienced their partner and children as a very important factor to keep fighting for their lives (28) and made them realise they still had high QoL (24). However, some studies showed that patients who felt more of a burden the more support they received (23; 28) had a lower QoL.

Some studies reported about how the daily lives of patients changed since the diagnosis. Some patients had to stop working abruptly (23; 24), others tried to keep their lives as normal as possible (21; 23). However, some changes could not be prevented, and patients had to adjust their lives. Hunting changed, for instance, to bird watching, and travelling was modified to watching travel documentaries. Apart from the physical activities, patients also experienced changes in their social contacts. The social network was of great importance to the patients, as this network helped them to accept their limitations and provide them with meaningfulness in their lives (31; 32). In some cases the ALS led to a loss of social contacts (28), resulting in loneliness and a decline in QoL.

*PPS*: Different aspects influence the QoL of people with PPS, most of them being related to participation in daily life. The first aspect or barrier concerned (physical) accessibility; many public places or public transportation are not accessible for people who cannot climb stairs, walk long distances or use a wheelchair (38). Feelings of exclusion due to the physical environment had negative consequences on the QoL.

A second aspect mentioned in the literature had to do with the attitudes of the social environment. Patients often felt they had to prove themselves and 'battle' to be accepted within an 'able-bodied' world (38). Related to this, different studies reported that patients with PPS could not rest enough and felt the urge to keep going, sometimes with the consequence of wearing themselves out (26; 39). Having polio as a child in the 1950s resulted in developing personality traits such as 'determination', 'optimism', or 'strong willed', 'bolshie' 'not asking for any compromises'. Patients felt that this helped them to succeed in life and become 'survivors' (38). This fighter-spirit sometimes led to difficulties with PPS; managing their occupations was one of the most difficult challenges for patients (39). Patients feared possible rejection by society; they were scared to be appreciated solely for their outer qualities such as health, beauty, and productivity (26). This seemed to influence their QoL in a negative way.

It is interesting to see that the QoL of patients does not correlate with the severity of the disease. For people with PPS, the QoL is much influenced by their possibility to take part in daily activities. This differs from the people with ALS, where the QoL seems to be determined by more social factors, such as receiving social support and being engaged in relations.

## **Coping**

A widely discussed theme in the articles was the coping of patients with ALS and PPS. For PPS, a separate study was written about different coping styles apart from the articles that paid indirect attention to coping. For ALS, coping styles were discussed more indirectly.

*ALS*: Different types of coping strategies can be distinguished; a) strategies grounded in a positive attitude; b) strategies based on denial; and c) strategies focussing on activities. The strategy grounded in a positive attitude was found in patients reporting that they 'decided' to 'live on the sunny side of the street' (24). Humour, teasing and black humour became active strategies to counteract inner negative feelings and demoralising aspects of the disease (27; 38). The style of denial was found among patients stating that they were not to complain, as others had even worse diseases (29). Ways of denying included dreaming of the improbable, affirming 'I'm doing all right' or 'giving in' to ALS (27), resulting in anger and sorrow (32). Another form that could be considered denial was patients stating that they only lived by the day (24; 27; 29; 32).

The third style was found in patients being very motivated to improve their own lives or the lives of others. This strategy was especially used by people who felt they lost control over their lives due to the ALS. This prompted them to exert control in health care (30). This control was not only sought-after in traditional medicine, but could also be found in alternative means (27; 29) or the urge to fight for the rights of the disabled (28). Often, these activities gave patients the feeling of being in control and being able to contribute to society (27; 28; 30; 35).

One study looked specifically at the storylines represented by patients with ALS. This study recognised four different storylines in the narratives of patients with ALS: diverging from a fracturing, preserving, enduring and sustaining one's storyline (29).

*PPS:* Many different coping strategies were described for people with PPS. In general, two different types of strategies can be distinguished: a) strategies based on acceptance and b) strategies based on denial. Strategies related to acceptance included, for example, adjusting behaviour and changing activities, such as setting priorities and giving up occupations, and planning ahead to avoid fatigue after such activities (26; 33; 38; 39). Seeking support, informing people about the disease and its consequences and looking for alternative therapies were also used by many patients (33; 38; 39). Strategies related to denial were taking life each day at a time, avoiding thinking about the disease and passing the limits of one's own body (26; 34; 39). Although the people with PPS used different coping strategies, coping with their daily disabilities was experienced as a difficult process (33; 38) .

Because many studies described coping strategies of patients with ALS and PPS, many different descriptions occurred about these strategies. One strategy that both patient groups seem to use is denial, where they try to keep going on the way they used to. For people with PPS this might be possible for a longer time, but sooner or later both patient groups have to give in to their increasing disabilities.

### **End of life decisions**

Several articles looked into the feelings of ALS patients considering their end of life. For PPS this theme was not mentioned in the articles, as it is probably not so urgent. Two themes related to end of life were important in ALS: being 'in control' and maintaining dignity.

*Being in control:* Different studies showed that being in control in the last phase of the disease is of great importance to the patients (30; 32). The studies showed that patients are often very open about their wishes regarding end of life. They had the desire to follow through with their own preferences regarding their end of life. Patients did, however, experience anxiety both from the journey towards death and, to a lesser

extent, from death itself (31). By being in control over their end of life, patients also tried to make their death easier for those who would survive them (31).

The amount of possible control over one's end of life differs per country, due to the different legislation towards euthanasia and assisted suicide. One study described the thoughts about euthanasia and suicide of the people with ALS. Patients who lived in countries where euthanasia was not allowed were more scared of death than people who could choose euthanasia. The issues of assisted suicide and euthanasia were still considered taboo in some countries in the context of ALS care (23). Not in all cases euthanasia or assisted suicide were mentioned as a condition of being in control. Patients also felt control in their decisions whether or not having a feeding tube or a tracheostomy (30). The ventilator was a way to feel in control of the end of life, as patients could decide whether they wanted to use it or not.

*Dignity:* Patients articulated that it was important for them to die with dignity(32), being able to breathe, having little pain, and being able to 'cross the line a bit earlier' (euthanize) (23). Patients feared becoming completely dependent regarding personal hygiene needs (27). In addition, not being able to move was feared by some patients (27). Some patients stated that they would not want to experience such dependency and would prefer the option of euthanasia, whereas others rejected the option of euthanasia. The fact that it is not possible for patients in some countries to be euthanized was frightening for them (23).

Issues considering end of life can be important for patients, as it gives them the feeling of being in control.

## DISCUSSION

This review highlights the illness experiences of patients with NMDs. Six themes emerged from the search: diagnosis, experience of symptoms, the use of devices, QoL, coping and end of life decisions. The themes showed that the experiences of the patients with NMDs were studied from the perspective of the medical and psychological paradigm. The focus was on the disease, its physical effects and the mental state of patients. This is shown, for example, by the studies that focused on the use of devices and coping. However, the medical and psychological paradigm is rather narrow, leaving little space for the role of the context the patient is part of. This context, however, can be an important factor influencing the experiences of people with NMDs (40; 41). The more recent studies did regularly take some social aspects into account. Here, a more critical approach of social structures and mechanisms of social exclusion were found.

Besides, some other salient points need to be discussed. First, the amount of studies among the diseases is noteworthy. None of the articles studied the illness experiences of patients with FSHD. Compared to the amount of ALS studies, the studies about PPS were limited (5 out of 20). This shows that there is more attention for ALS than for PPS or FSHD. The reason for this focus on ALS is unknown to us. A possible explanation could be that FSHD and PPS were not specifically addressed in a study, but rather described in more general studies about muscular dystrophy (for FSHD) or motor neuron disorders (for PPS). Some articles were published about these, more general, disorders (42; 43), but this does not seem to entirely explain the difference. The emphasis on ALS could also originate from the fact that, contrary to FSHD and PPS, ALS is a rapidly progressive disease that leads to death often within a couple of years. This rapid progression and fatal character of the disease force patients and health care professionals to anticipate on future declines and closely mirror the deterioration. This constant awareness of the deterioration could lead to more responsiveness of the patient perspective. Another possible explanation is that fatal diseases like those of ALS and cancer attract societal and media attention, and generate more money for research from charities and local actions. As most studies in our review concentrated on the quick progression of ALS, this might have led to the dominance of the medical model. There was lesser attention for the social context of the increasing disablement, other symptoms were deemed more urgent. The acute nature of ALS and its progression might explain the fact that most studies about ALS were discussed from a medical and psychological paradigm, with less attention for the broader context of the social and personal consequences of living with this disease.

Being in control during the illness process was one of the main themes, mentioned in many of the 20 articles. ALS and PPS patients experienced increased dependency as negative, sometimes resulting in feelings of shame, guilt, sorrow or anger. The loss of independency can lead to feelings of loss of identity as participation in everyday life and activities is less straightforward. Becoming more dependent often means losing autonomy. This specific loss of autonomy is difficult in the Western society, where precisely individualism and autonomy are the guiding principles (44-48). The perfect body in an individualistic society is a body of a physically perfect person, without weakness, loss or pain. (49; 50). This model of the perfect body implies that people are supposed to control their bodies (50; 51). It should be noted that the model of the perfect body differed between people with PPS and people with ALS. For people with PPS, autonomy and independence has often been a great issue throughout their lives; they often fought for independence in their childhood, adolescence and early adulthood, and became used to their altered body. Former studies address the fact that the timing of impairment in the life course may influence how it impacts people's lives (52; 53). Despite this difference in illness history, the loss of independence and autonomy applies to both people with PPS and people with ALS. Fact is that not being

(fully) in control, therefore a great impact on the illness experiences of people in our Western society.

In the context of a individualism, it is remarkable that relatively little attention is paid on the use of devices, as devices are ideals tools to improve peoples' autonomy. Patients with PPS and ALS often have to use devices such as canes, walkers, wheelchairs, or speaking devices. Other studies showed that the use of devices had a positive as well as negative impact on patients' lives (44; 54). On one hand, using a device can affect social status and identity and can lead to possible social stigmatization (44; 54; 55). On the other hand, the use of a device can also improve social participation, as it compensates for the impairments that eventually can lead to a more independent everyday life (44; 56). In any case, the use of devices influences patients' lives, but this impact has barely been studied in the articles included in this systematic review. An exception is the attention for the use of ventilation for people with ALS. This exception might be explained by the fact that most studies included in this review focused on ALS. However, the experiences with these devices are also described as factual; lesser attention is paid to the experiences of patients who are dependent on a ventilator.

### **Implications for rehabilitation**

In rehabilitation care, the overarching aim is to enable people to participate in society. Traditionally, rehabilitation focused mainly on the improvement of physical aspects of functioning and adaptations to compensate activity restrictions due to lasting impairments, in order to achieve societal participation. Over the last decades, rehabilitation has shifted to a more context-related care, where physical, as well as social aspects of rehabilitation are addressed. In the perspective of this more holistic approach, illness experiences have become increasingly important. The personal circumstances of people, and correspondingly their illness experiences, can play a great role in how a diagnosis and treatment are experienced. This means that illness experiences can give insight in what is important for people at certain points in their lives and, in line with this, in health care practice. Taking these illness experiences and perceptions into account in rehabilitation care can therefore both be fruitful in achieving better outcomes regarding societal participation (10) as well as enabling a better aligned, patient-centred health care practice.

We should be aware that most studies of this review were dedicated to experiences of ALS. This shows a lack of understanding considering illness experiences in other NMDs. Thus, we recommend more research on illness experiences in other NMDs to understand the impact of the disease and therefore the consequences for rehabilitation care.

## Methodological considerations

There are limitations considering the inclusion criteria, search process, analysis, and positioning of the reviewers of this study. This review only considered articles published in peer reviewed scientific journals written in English. Therefore, there was an inherent publication bias excluding other potential sources of evidence such as grey literature or research published in books (57). The fact that this review only identified 20 studies that met the inclusion criteria, does not, therefore, necessarily signify a dearth of relevant research or publications.

Effort was taken to enclose all relevant articles in the search; various synonyms for the diseases and illness experiences were used; we searched not only titles but also abstracts for relevant concepts. However, the use of inappropriate or inconsistent terminology and indexing of research could have resulted in the omission of relevant studies (57; 58).

As this review only focused on the patients' perspective, the viewpoint of the caregiver was not taken into account even though their experiences can also be of value. However, because the objective was to gain insight on the patient's perspective, they are the most reliable informants regarding their own experiences (20). In order to gain a full understanding of the impact of a disease, it might be interesting to include the caregivers' perspective in future research.

A last methodological consideration is the fact that we have chosen ALS, PPS and FSHD as representative for the broader spectrum of NMDs. Even though these three types were chosen carefully and with great consideration, the fact that they are representatives for a larger group is a limitation of this study as it is probable that every separate illness has its own problems. However, recent research showed overlap in the experiences of people with different NMDs (59). Therefore, it is assumed that the combination of these NDMs already give a broader insight in the illness experiences than the selection of one specific disease.

## CONCLUSION

This systematic review gives an overview of the existing studies on illness experiences in NMDs. Relevant themes such as experience with diagnosis, experience with symptoms, Quality of Life, use of devices, coping, decisions concerning end of life give an impression of factors that play an important role in the lives of patients with ALS and PPS. These themes show that the current literature is focused on the more biomedical and psychological aspects of the illness experiences, such as coping and quality of life. Little attention is paid to the broader context of living with a NMD, such as aspects of social inclusion and exclusion.

The content of the six themes differ between ALS and PPS; aspects as use of devices, quality of life and experiences with symptoms are at least partly determined by the history of the disease. In PPS people have often lived their entire lives with sequelae of the acute polio, whereas people with ALS have lived in good health for the best part of their life. It is not surprising that this results in differences between ALS and PPS. However, the results of this study also show that, despite the differences, both illnesses partly deal with the same issues, such as loss of independency and autonomy and also coping strategies show similarities.

## DECLARATION OF INTEREST

The authors report no declarations of interest.

## ACKNOWLEDGEMENTS

The study was supported by grants from the Netherlands Organisation for Health Research and Development (ZonMw) and 'Prinses Beatrix Spierfonds'.

## APPENDIX

### Search strategy in PubMed

Search	Query	Results
#7	#3 AND #6	1158
#6	#4 OR #5	606161
#5	Life change[tiab] OR (adapt*[tiab] AND psycholog*[tiab]) OR coping[tiab] OR emotion*[tiab] OR mood*[tiab] OR quality of life[tiab] OR illness experience*[tiab] OR illness perception*[tiab] OR subjective experience*[tiab] OR illness narrative*[tiab] OR (Illness[tiab] AND experienc*[tiab]) OR (Illness[tiab] AND perception*[tiab]) OR (Illness[tiab] AND narrati*[tiab])	376430
#4	("Life Change Events"[Mesh] OR "Sick Role"[Mesh] OR "Adaptation, Psychological"[Mesh] OR "Emotions"[Mesh] OR "Sickness Impact Profile"[Mesh] OR "Quality of Life"[Mesh])	389685
#3	#1 OR #2	24142
#2	ALS[tiab] OR Amyotrophic Lateral Sclerosis[tiab] OR Postpoliomyeliti*[tiab] OR post-polio*[tiab] OR Fascioscapulohumeral*[tiab] OR Facioscapulohumeral*[tiab] OR Fascio-scapul*[tiab] OR Facio-scapul*[tiab]	21750
#1	"Amyotrophic Lateral Sclerosis"[Mesh] OR "Postpoliomyelitis Syndrome"[Mesh] OR "Muscular Dystrophy, Facioscapulohumeral"[Mesh]	14628

### Search strategy in Embase.com

Search	Query	Results
#7	#3 AND #6	3349
#6	#4 OR #5	918184
#5	'life change':ab,ti OR (adapt*:ab,ti AND psycholog*:ab,ti) OR coping:ab,ti OR emotion*:ab,ti OR mood*:ab,ti OR 'quality of life':ab,ti OR (illness NEXT/1 experience*):ab,ti OR (illness NEXT/1 perception*):ab,ti OR (subjective NEXT/1 experience*):ab,ti OR (illness NEXT/1 narrative*):ab,ti OR (illness:ab,ti AND experienc*:ab,ti) OR (illness:ab,ti AND perception*:ab,ti) OR (illness:ab,ti AND narrati*:ab,ti)	514609
#4	'life event'/exp OR 'adaptive behavior'/exp OR 'emotion'/exp OR 'sickness impact profile'/exp OR 'quality of life'/exp	723714
#3	#1 OR #2	70017
#2	als:ab,ti OR 'amyotrophic lateral sclerosis':ab,ti OR postpoliomyeliti*:ab,ti OR (post NEXT/1 polio*):ab,ti OR fascioscapulohumeral*:ab,ti OR fascioscapulohumeral*:ab,ti OR (fascio NEXT/1 scapul*):ab,ti OR (facio NEXT/1 scapul*):ab,ti	63187
#1	'amyotrophic lateral sclerosis'/exp OR 'postpoliomyelitis syndrome'/exp OR 'fascioscapulohumeral muscular dystrophy'/exp	27006

### Search strategy in PsycINFO (via EBSCO)

Search	Query	Results
S7	S3 AND S6	1044
S6	S4 OR S5	529650
S5	TI ( "Life change" OR (adapt* AND psycholog*) OR coping OR emotion* OR mood* OR "quality of life" OR "illness experience*" OR "illness perception*" OR "subjective experience*" OR "illness narrative*" OR (Illness AND experienc*) OR (Illness AND perception*) OR (Illness AND narrati*) ) OR AB ( "Life change" OR (adapt* AND psycholog*) OR coping OR emotion* OR mood* OR "quality of life" OR "illness experience*" OR "illness perception*" OR "subjective experience*" OR "illness narrative*" OR (Illness AND experienc*) OR (Illness AND perception*) OR (Illness AND narrati*) )	388615
S4	(((((((DE "Life Changes") OR (DE "Adaptive Behavior") OR (DE "Adjustment")) OR (DE "Emotional Adjustment")) AND (DE "Emotional Control" OR DE "Anger Control")) OR (DE "Identity Crisis")) OR (DE "Emotions")) OR (DE "Emotional States" OR DE "Affection" OR DE "Alienation" OR DE "Ambivalence" OR DE "Anger" OR DE "Anxiety" OR DE "Apathy" OR DE "Aversion" OR DE "Boredom" OR DE "Depression (Emotion)" OR DE "Disappointment" OR DE "Disgust" OR DE "Dissatisfaction" OR DE "Distress" OR DE "Doubt" OR DE "Embarrassment" OR DE "Emotional Trauma" OR DE "Enthusiasm" OR DE "Euphoria" OR DE "Fear" OR DE "Frustration" OR DE "Gratitude" OR DE "Grief" OR DE "Guilt" OR DE "Happiness" OR DE "Helplessness" OR DE "Homesickness" OR DE "Hope" OR DE "Hopelessness" OR DE "Jealousy" OR DE "Loneliness" OR DE "Love" OR DE "Mania" OR DE "Mental Confusion" OR DE "Optimism" OR DE "Pessimism" OR DE "Pleasure" OR DE "Pride" OR DE "Regret" OR DE "Restlessness" OR DE "Sadness" OR DE "Shame" OR DE "Suffering" OR DE "Suspicion" OR DE "Sympathy")) OR (DE "Quality of Life")	255838
S3	S1 OR S2	8528

S2	TI ( (ALS OR "Amyotrophic Lateral Sclerosis" OR Postpoliomyeliti* OR post-polio* OR Fascioscapulohumeral* OR Facioscapulohumeral* OR Fascio-scapul* OR Facio-scapul*) ) OR AB ( (ALS OR "Amyotrophic Lateral Sclerosis" OR Postpoliomyeliti* OR post-polio* OR Fascioscapulohumeral* OR Fascioscapulohumeral* OR Fascio-scapul* OR Facio-scapul*) )	8468
S1	DE "Amyotrophic Lateral Sclerosis"	2374

### Search strategy in CINAHL (via EBSCO)

Search	Query	Results
S7	S3 AND S6	417
S6	S4 OR S5	193098
S5	TI ( "Life change" OR (adapt* AND psycholog*) OR coping OR emotion* OR mood* OR "quality of life" OR "illness experience*" OR "illness perception*" OR "subjective experience*" OR "illness narrative*" OR (Illness AND experienc*) OR (Illness AND perception*) OR (Illness AND narrati*) ) OR AB ( "Life change" OR (adapt* AND psycholog*) OR coping OR emotion* OR mood* OR "quality of life" OR "illness experience*" OR "illness perception*" OR "subjective experience*" OR "illness narrative*" OR (Illness AND experienc*) OR (Illness AND perception*) OR (Illness AND narrati*) )	104847
S4	(MH "Life Change Events+") OR (MH "Sick Role+") OR (MH "Adaptation, Psychological+") OR (MH "Emotions+") OR (MH "Sickness Impact Profile+") OR (MH "Quality of Life+")	132185
S3	S1 OR S2	3049
S2	TI ( (ALS OR "Amyotrophic Lateral Sclerosis" OR Postpoliomyeliti* OR post-polio* OR Fascioscapulohumeral* OR Facioscapulohumeral* OR Fascio-scapul* OR Facio-scapul*) ) OR AB ( (ALS OR "Amyotrophic Lateral Sclerosis" OR Postpoliomyeliti* OR post-polio* OR Fascioscapulohumeral* OR Facioscapulohumeral* OR Fascio-scapul* OR Facio-scapul*) )	2315
S1	(MH "Amyotrophic Lateral Sclerosis+") OR (MH "Postpoliomyelitis Syndrome+") OR (MH "Muscular Dystrophy, Facioscapulohumeral+")	2293

### Search strategy in COCHRANE LIBRARY (via Wiley)

Search	Query	Results
S1	(ALS OR "Amyotrophic Lateral Sclerosis" OR Postpoliomyeliti* OR post-polio* OR Fascioscapulohumeral* OR Facioscapulohumeral* OR Fascio-scapul* OR Facio-scapul*):ti,ab,kw AND ( "Life change" OR (adapt* AND psycholog*) OR coping OR emotion* OR mood* OR "quality of life" OR "illness experience*" OR "illness perception*" OR "subjective experience*" OR "illness narrative*" OR (Illness AND experienc*) OR (Illness AND perception*) OR (Illness AND narrati*) ):ti,ab,kw	143

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