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A QUALITATIVE SYSTEMATIC REVIEW OF LIVING WITH DUCHENNE MUSCULAR DYSTROPHY

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Abstract

Duchenne muscular dystrophy (DMD) is one of the most severe forms of inherited muscular dystrophies, leading to limited upper limb use in its later stages. Due to medical and technological advances, patients with DMD are living longer and are reaching adolescence and adulthood. The benefits of bringing together qualitative views and studies in a systematic way include gaining a greater breadth of perspectives and a deeper understanding of health, psychological issues, and concerns from the point of view of patients, and helping *the medical community and policymakers to create a better support system. This review aimed to describe the experiences of patients living with DMD in order to identify common themes across all relevant studies. The research question is: What is the lived-world experience of men with DMD? This review adopted a systematic search methodology in accordance with PRISMA guidance, and followed the SPIDER search strategy and the analytic procedure for thematic synthesis.*

In total, 11 studies were included in the review concerning 132 young people with DMD. In these studies, 5 main topics were identified: experiences of body powerlessness (progressive loss of body control, limited social freedom, lack of privacy); the search for identity (the meaning of becoming an adult man, feelings at the margins, maintaining normality, balancing dependence-independence); challenges in social relations (vulnerability to prejudice, risk of isolation, (in)dispensable parental support, emotional connectedness with carers, the support of others "in the same boat" with DMD); coping processes and resilience (underestimated strength, taking autonomy, toward acceptance, active self-management); and end-of-life experiences (living in the present, experiences of dying, (mis)communication with others about death and dying).

Keywords: *neuromuscular disease, progressive illness, male gender, qualitative methodology, thematic synthesis.*

Introduction

Duchenne muscular dystrophy (DMD) is a genetic disorder that occurs primarily in males, and is one of the most severe forms of inherited muscular dystrophies (Glover et al., 2020). The disease progresses rapidly and makes muscles weaker and less flexible (Angelini, 2018), leading to a loss of ambulation in childhood and limited upper limb use in its later stages. Patients are typically confined to a wheelchair by 12 years of age (Venugopal & Pavlakis, 2020). There is no cure for DMD and death occurs because of respiratory or cardiac complications (Venugopal & Pavlakis, 2020). The medical and technological advances in medical care have prolonged life expectancy into the third and sometimes fourth decade (Landfeldt et al., 2015). Children and young adults with DMD require the assistance of parents and close family members with all aspects of daily living as the condition progresses (Pangalila et al., 2012).

Due to medical and technological advances, young people with this condition that would previously have been limited to childhood are now living longer, and are therefore experiencing significant changes in their personal development as they undergo adolescence and transition into adulthood. It is essential to understand how they perceive and cope with this disease and the needs and concerns that they encounter. It is also important to understand the lived-world experience of men with DMD to help the medical community and policymakers to create a better support system.

The benefits of bringing together qualitative views and studies in a systematic way include gaining a greater breadth of perspectives and a deeper understanding of health, psychological issues, and concerns from the point of view of patients (Harden et al., 2004).

The objective of this study is: A systematic review of qualitative studies reporting the experience of patients living with DMD in order to identify common themes across all relevant studies.

1. Methods

This review adopted a systematic search methodology in accordance with PRISMA guidance (Moher et al., 2009) and followed the SPIDER search strategy (Cooke et al., 2012) and the analytic procedure for thematic synthesis (Thomas & Harden, 2008).

Search strategy. A search of literature was conducted using multiple databases, including: PubMed, Web of Science, EBSCOhost, and Sage Journals. Reference lists from relevant papers were hand searched, and all databases were searched between June and August 2018. The search terms and the final search strategy were developed using the SPIDER (Sample, Phenomenon of Interest, Design, Evaluation, Research type) search tool (Cooke et al., 2012), which is specifically designed for qualitative evidence synthesis. Details of the search terms and the search syntax are given in Table 1.

Table 1. *Database search strategy*

Sample	**child, adolescent, teen, young people, men**
Phenomenon of Interest	Duchenne muscular dystrophy
Design	interview
Evaluation	view, experience, reflection, attitude, perception, beliefs, feelings
Research type	qualitative
Final search strategy S AND PI AND (D OR E OR R)	(child* OR adolescent* OR teen* OR young people OR men*) AND Duchenne muscular dystrophy AND (interview* OR view* OR experienc* OR reflect* OR attitude* OR perce* OR belie* OR feel* OR qualitative)

Eligibility criteria. Articles were considered for inclusion if they were qualitative studies written in English that explored the experience of boys or young men living with DMD. Quantitative and mixed method studies, literature reviews, studies involving groups with multiple illnesses, parents, siblings, or specific DMD treatments, and non-English language papers were excluded. Studies published prior to 1990 also were excluded due to progress in the diagnostic approaches to and management of DMD with steroids and ventilation over the past decades (Lynn et al., 2015), as this may have altered the perceptions of the illness (Glover et al., 2020). All inclusion and exclusion criteria are given in Table 2.

Table 2. *Inclusion and exclusion criteria*

	Inclusion criteria	**Exclusion criteria**
Study design	Qualitative methodology, primary research	Quantitative methodology, mixed methods, reviews of the literature
Population	Children or men with DMD	Studies involving groups with multiple illnesses Studies involving children or men with DMD and their parents or siblings
Study focus	Children, men experiences of living with DMD	Focus on specific treatment. Focus on patient family, parents, siblings
Date	1990–2018	Studies published prior to 1990
Language	English language papers	Non-English language papers

Data extraction. The screening process involved several stages. First, the databases were searched and the duplicates removed. Then the titles of the papers were reviewed, and where the focus was not clear in the title, the abstract was examined. When the abstract was not descriptive enough, the full text was examined. Sources not fulfilling the inclusion criteria were excluded. For each included study, the following data were extracted: author, year, country, purpose of study, data collection strategy, data analysis, sample, and main findings. As well as basic study information, all participant quotations and text presented within the findings/results or conclusion/discussion sections of each included article were extracted. This included data as expressed by the participants and by the authors.

Data analysis. For data analysis, the method of thematic synthesis described by Thomas and Harden (2008) was used. The purpose of this method is to develop analytical themes through a descriptive synthesis and find explanations relevant to a particular review question. It provides a systematic and transparent approach which facilitates the production of rich, interpretive, yet data-driven themes in order to broaden conceptual understandings of a particular phenomenon (Heath et al., 2017). The stages of thematic analysis are (Thomas & Harden, 2008): 1) free line-by-line coding of textual findings from primary studies; 2) organization of free codes into descriptive themes; and 3) generation of analytical themes. While the development of descriptive themes remains close to primary studies, analytical themes represent a stage of interpretation whereby the reviewers go beyond primary studies and produce a new interpretation (Heath et al., 2017).

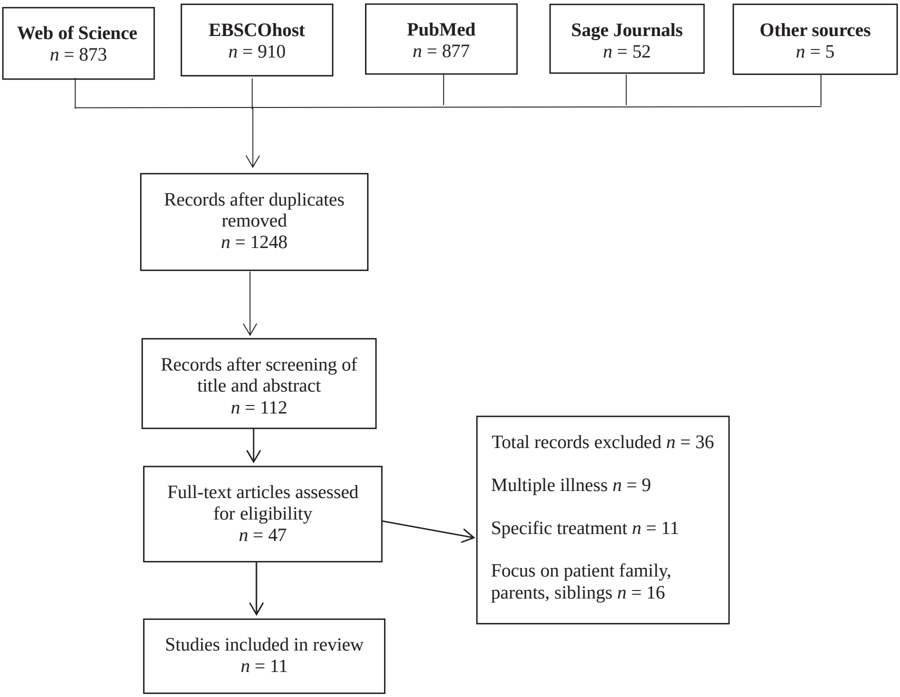

Figure 1. *A flow chart of the article selection and review process*

Quality appraisal. There is currently no consensus on how or whether to use quality appraisal in a qualitative synthesis (Dixon-Woods et al., 2004). In this review, critical appraisal was not used to exclude articles, but the reported methodology regarding included papers was appraised to understand any limitations. The studies were assessed using criteria reported by Thomas and Harden (2008). As seen in Table 3, the quality of the studies was generally high, although several studies were found to be lacking in one or more areas of reporting.

Table 3. *Quality appraisal of studies*

	Aims clearly reported	Adequate reporting of sample	Adequate reporting of data collection methods	Adequate reporting of data analysis methods	Appropriate research design	Appropriate recruitment strategy	Ethical issues considered
Gibson et al., 2007	Yes	Yes	Yes	-	Yes	Yes	Yes
Gibson et al., 2009	Yes	Yes	-	Yes	Yes	-	Yes
Pehler & Craft-Rosenberg, 2009	Yes	Yes	Yes	Yes	Yes	Yes	Yes

	Aims clearly reported	Adequate reporting of sample	Adequate reporting of data collection methods	Adequate reporting of data analysis methods	Appropriate research design	Appropriate recruitment strategy	Ethical issues considered
Dreyer et al., 2010	Yes	Yes	Yes	Yes	-	Yes	Yes
Yamaguchi & Suzuki, 2013	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Gibson et al., 2014	Yes	No	Yes	Yes	Yes	Yes	Yes
Abbott et al., 2016	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Fujino et al., 2016	Yes	Yes	Yes	Yes	Yes	-	Yes
Abbott et al., 2017	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Skyrme, 2017	Yes	No	Yes	No	Yes	Yes	-
Finkelstein & Marcus, 2018	Yes	Yes	Yes	-	Yes	Yes	Yes

- data not available

2. Results

The search yielded 2,717 papers. The full texts of 47 papers were retrieved and reviewed, and 11 studies met the inclusion criteria and were included in the review. Studies were conducted in six countries: the UK ($n = 3$), Canada ($n = 3$), Japan ($n = 2$), the USA ($n = 1$), Denmark ($n = 1$), and Israel ($n = 1$). The total number of young people with DMD consulted in these studies was 132, with ages ranging from 10 to 48 years. Of these participants, 50 lived with their parents in a family home, 41 in their own home or in a supportive housing apartment, and 6 in a long-term care facility. Three studies did not provide information on the living situation of respondents. Consistent with the progression of DMD, all respondents used wheelchairs for mobility, and 75 received ventilation via tracheotomies or facemasks. More details of the included studies are provided in Table 4.

Table 4. *Details of included studies*

Nr.	Study	Country	Objective	Data collection and analysis	Sample	Main findings
1.	Gibson et al., 2007	Canada	To explore the identities and social positionings of men with DMD in relation to dominant discourses of disability, masculinity and technology	Semi-structured interviews, video diaries. Ethnographic study. The data analysis described by Miles and Huberman (1994).	10 men with DMD aged 22–36.	Men with DMD were materially, socially, and symbolically marginalized through inaccessible built environments, social arrangements that limited their engagement in community life, and the multiple ways that their bodies were negatively marked across social space. Furthermore, their marginalization had been embodied through processes of socialization and the internalization of subordinate social positions.
2.	Gibson et al., 2009	Canada	To examine the effects of a shifting life expectancy on personal identities.	Interviews. Coding system.	10 men with DMD aged 22–36.	Participants' narrative accounts revealed how their dispositions were orientated to a shortened lifespan that exerted damaging effects regardless of actual lifespan. Compounding their material, social, and symbolic isolation was a temporal isolation whereby the men had lived every day anticipating that it could be their last for as much as a decade.
3.	Pehler & Craft-Rosenberg, 2009	USA	To describe the lived experiences of spirituality in adolescents with DMD.	Interviews. Van Manen's Phenomenological method.	9 male adolescents with DMD aged 12–17.	The essential theme of spirituality was "longing" – the strong desire for something unattainable. For understanding spirituality, participants mediated their longing through "connecting with others, self, and beyond self."
4.	Dreyer et al., 2010	Denmark	To study the life-experiences of people living with DMD, home mechanical ventilation (HMV), and physical impairment.	Narrative interviews. Phenomenological hermeneutic approach, analysis inspired by Ricoeur.	19 patients with DMD and invasive HMV aged 21–40.	HMV not only extended the participants' lifespans; it also gave them the capacity to live an active life. They were totally dependent in everyday living, but, in spite of this, they did not see themselves as physically impaired. They realized that there were activities that were physically impossible, but they considered themselves to be just the same person they had always been. This dependency was described as "independent dependency."

Nr.	Study	Country	Objective	Data collection and analysis	Sample	Main findings
5.	Yamaguchi & Suzuki, 2013	Japan	To better understand the process by which individuals with DMD arrived at goal for independence (i.e., choosing to live at home in Japan instead of in special sanatoriums that provide sufficient support and care).	Semi-structured interviews. Grounded theory.	21 men with DMD (mean age 31.4 ± 10.8).	The core element underlying participants' goals for independent living was self-reliant independency. To improve their social inclusion, the strategies used by the participants to retain their autonomy in an underdeveloped Japanese welfare system by establishing relationships with people in their communities can prevent them from experiencing social isolation.
6.	Gibson et al., 2014	Canada	To investigate the intersectionality of gender, disability, and emerging adulthood with young men with DMD.	Interviews, photo-elicitation, and solicited narrative diaries. Inductive analyses identify new themes.	11 young men with DMD, aged 16–27.	Analysis suggested that disability, masculinities, and generational (life stage) identities intersected through "narratives of nondifference," wherein participants worked to establish identities as typical "guys." Within limited fields of school and work, participants distanced themselves from the label of "disabled" and discussed their successes and challenges in terms of normative developmental trajectories.
7.	Abbott et al., 2016	UK	To explore men's experiences of the organization and delivery of social care as it pertained to their sense of being men.	Interviews. Thematic analysis.	20 men with DMD aged 21–33.	Social care in its broadest sense did little to support a positive sense of masculinity or male gender. More often than not, the organization and delivery of social care people de-gendered or emasculated many of the men who took part in the study.
8.	Fujino et al., 2016	Japan	To examine the experiences of patients with DMD in facing and learning about their disease.	In-depth interviews. Thematic analysis.	7 men with DMD aged 20–48.	Main themes: 1) experiences before receiving the diagnosis; 2) experiences when they learned of their condition and progression of the disease; 3) support; and 4) desired explanations. Anxiety and worry were most pronounced when respondents had to transition to using wheelchairs or respirators due to disease progression; indeed, such transitions affected the patient's psychological adjustment.

Nr.	Study	Country	Objective	Data collection and analysis	Sample	Main findings
9.	Abbott et al., 2017	UK	To focus on the views and preferences of men with DMD and how they could best be supported to approach end of life planning.	Semi-structured interviews. Thematic analysis.	15 men with DMD aged 20–45.	Topics of interest to participants included likely nature and place of death; practical planning for funerals and wills; and sources of information and support. Emotional or psychological support to think about end of life was not routinely offered, and participants found it very difficult to discuss these issues with family members.
10.	Skyrme, 2017	UK	To discuss aspects of the participants’ lives: to talk about the impact of discomfort; declining mobility; and social issues such as discrimination and a sense of difference.	Semi-structured interviews, imaginative scenarios.	9 boys aged 10–21.	Living with a degenerative condition has physical and social impacts, and there are limited opportunities for patients to talk about these issues. It was also apparent that parents are key figures who provide care for their child, often in the absence of comprehensive support services.
11.	Finkelstein & Marcus, 2018	Israel	To learn directly from an adult person with DMD about how he experiences independence given his total dependence on people and technology.	In-depth open interviews. Phenomenological paradigm.	Eytan (aged 44).	Two main topics stand out. The first was Eytan’s relations with his caretakers. This study shows how essential Eytan’s relationships with his caretakers were to his quality of life. The second topic was what Eytan considered to be “a life worth living” – namely his relations with his family and friends. Eytan placed a high value on autonomy, but his experience shows that this may be achieved through social relationships involving decisions, actions, and people in ways that could be mistaken for signs of non-autonomy.

Within the included papers, 5 main topics were identified: experiences of body powerlessness, the search for identity, challenges in social relations, coping processes and resilience, and end-of-life experiences.

2.1. Experiences of body powerlessness

This topic includes 3 subthemes: progressive loss of body control (1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11), limited social freedom (2, 3, 4, 5, 6, 10), and lack of privacy (7, 11).

Progressive loss of body control. The pathogenesis of the disease itself suggests that muscle weakness is a major symptom of DMD. However, there is evidence that patients with DMD have a wide range of physical complaints, from general fatigue to lung problems and headaches. All 11 cases analyzed included features of loss of body control. It is important to understand that being in a state of non-fulfilled body control can affect day-to-day functioning; for example, fatigue can cause attention problems. Despite the progression of general loss of body control, some bodily functions do not deteriorate. For example, the smooth muscles responsible for the contraction of hollow organs such as blood vessels or the intestines remain unchanged. On the other hand, the risk of being overweight can be a real problem as patients have forced sedentary lifestyles and move less.

Loss of body control can be not only objective but also subjective; it is connected with the level of brain function, especially cognitive abilities. This is due to the fact that protein dystrophin is present not only in muscle tissues but also in the central nervous system. Thus, cerebral dystrophin deficiency may affect cognitive activity in boys. The normal dystrophin isoforms are distributed in hippocampal and purkinje cells in the cerebellum, contributing to higher postsynaptic density. This altered postsynaptic plasticity can disrupt efficient memory, automation, and planning.

Limited social freedom. Physical disability in individuals with DMD causes difficulties in communicating both directly and indirectly. As the ability to control the body weakens, the environment becomes filled with obstacles. Things that used to cause no problem in performing daily activities, and that were not even worth thinking about, start to become impossible. The presence of stairs and unsuitable transport increasingly hinders various activities, including leaving the house, meeting friends, and studying. From an early age, boys with DMD have difficulty finding friends or maintaining existing friendships as their symptoms worsen, as children often spend time running, jumping, cycling, playing sports, and so on (3, 4, 10). These children, who do not have the opportunity to engage in similar physical activities, often spend less and less time together and feel excluded, isolated, lonely, angry, and different from others (1, 3, 4).

In adolescence, these problems are exacerbated and become particularly significant when young men are often unable to spend time with others and participate in friends' gatherings (3, 4). In such a situation, the use of the Internet can reduce separation (4). As the condition worsens, it takes an increasing amount of planning to leave home, and the help of others is needed (1, 10). Physical disability also reduces the chances of experiencing intimacy, making it even more difficult to make close contact and to find a girlfriend (4). Motivation to study and to work on the individual's education varies, both due to difficulties in going to school and because young people with DMD often perceive it as futile (2, 3, 4, 5, 6).

Lack of privacy. In living with DMD, the lack of privacy and the demonstration of nudity is daily and fundamental, especially in the later stages of the disease. Men who are unable to move are forced to accept help, without which they would not be able to look after themselves. They are fed, rearranged, washed, etc., by carers. Some men claim to be accustomed to a lack of privacy as the disease progresses slowly (11), while others experience it sensitively and try to actively deal with it (7). One study observed that in order to experience sexual satisfaction, other individuals must also be involved when the functions of the hands are numb, and the person is no longer able to masturbate (7).

Here we see that all participants have gone through the rapid and intense experience of body powerlessness. Patients reported this as a frustrating or traumatic experience, causing hopelessness and devastation due to the uncontrollable nature of the disease, as until recently all pharmacological attempts to stop the disease were fruitless. These data highlight important implications for these patients, which are not only physical but also psychological in nature.

2.2. The search for identity

This theme includes 4 subthemes: the meaning of becoming an adult man (1, 2, 4, 5, 6, 7, 10, 11), feelings at the margins (1, 3, 6, 7, 8, 9, 10, 11), maintaining normality (1, 3, 4, 6, 7, 8), and balancing dependence-independence (1, 4, 5, 7, 10, 11).

The meaning of becoming an adult man. An important topic revealed by researchers was the meaning of becoming an adult man, which considers what it means for an adolescent male to become an adult when they grow older. This topic is often accompanied by an analysis of masculinity, and is related to stereotypes of masculinity in society (7, 2, 10). When growing older, boys face various challenges in adhering to the norms of social life; in their case, the direct trajectory of adult life – work, wife, children, etc. – is no longer visible.

One of the most critical topics in this regard is sexuality and intimate relationships. Many participants had concerns about their attractiveness and body image, which could challenge their senses of themselves as men (7). It was relatively common for participants to reject fields such as dating, work, or school because they felt that they could not functionally participate in these fields (1). Men tended to associate the transition to adulthood with moving out of their parents' home and giving up parental care (1, 2, 6). By thus becoming independent, living for themselves, and no longer being as comfortable as they were with their parents, they began to "be in the world" (2). Another aspect of adult life is self-awareness, acceptance, and the perception of one's condition as it is, along with accepting other challenges such as the growing need for help from others and progressive disability (4, 5, 6, 7, 10).

Feelings at the margins. Adolescents and men experience a range of different emotions, but often tend to keep them to themselves (3, 6, 7, 9, 10, 11). On the other hand, it is also difficult for those around them to talk about their feelings (7, 8, 9). Some studies with slightly older young people with DMD found that men have a predominant stereotype of masculinity that is sought to be maintained by "not mentioning negative feelings" (6, 7, 11). However, internally men have a variety of fears, especially those that relate to their future death (9) – whether distant or near. Men with DMD also often experience anger at their exclusivity and condition (8), sadness, or a longing for the past (3).

Maintaining normality. The theme of being "normal" is often emphasized in DMD studies (1, 3, 4, 6, 7). Adolescents and men with DMD need to be seen as normal – like everybody else (1, 4, 6, 7). Although physical disability is the distinguishing external feature of "otherness" to those around it, people with DMD did not feel that way. They saw themselves as regular, typical men (1, 6, 7): "I see it when I look in the mirror, but I do not picture myself that way in my head; in my head, I am a normal person like everybody else" (4). Their sense of normalcy was supported and confirmed by the immediate social environment – families and friends (3, 4, 6).

Some participants worked to establish themselves as normal in relation to (usually nondisabled) peers (6). They took many strategies to establish that they were "just like everyone else," including "successes in school and plans for work, independence, engagement with sports as players or fans, and friendships" (1, 4, 6). In this way, they worked within their accounts to minimize or distance disease and disability from identity. For example, one participant said: "I don't really like it when people use the word 'disability.' Disability sounds as if a person is broken. But people with DMD are not broken" (6). Some participants also distanced themselves from other disabled youth (1, 6) by describing nondisabled friends as being "more normal" than disabled friends, and thought that other disabled people had negative attitudes, were not working as hard as they could, and had "given up" (6).

Balancing dependence-independence. While growing up, participants gradually require more physical assistance. They understand that the only way to live with DMD is to be dependent on others (5). The more the disease progresses, and the more dependent they become, the more strongly they tend to pursue autonomy and independence (1, 4, 5, 7, 10, 11). Participants try to maintain independence in a variety of ways, including: performing as many independent actions as possible (1, 4, 5, 7, 10, 11); engaging in virtual sports or online educational training (4); making various decisions autonomously (1, 7, 11); and moving out of their parents' home. Some men felt that living independently with personal assistants maximized their sense of control (7), while on the contrary others "have tried dehospitalization and returned home despite complete or almost-complete physical dependence" (5). People with poorer conditions often tried to distinguish between physical dependence and psychological dependence. For example, one paralyzed participant "sees himself as independent because he has the authority to make decisions regarding his care, while the staff is only a machine whose role is to carry out his instructions" (11). In another study, one person tried to maintain independence by being on his own for a few minutes a day (1), while another achieved states of independence in his mind and mindset by cultivating self-confidence and achieving goals (7).

2.3. Challenges in social relations

This topic includes 5 subthemes: vulnerability to prejudice (1, 3, 4, 6, 7, 8, 10), risk of isolation (3, 4, 8, 9, 10, 11), (in)dispensable parental support (3, 4, 7, 8, 9, 10), emotional connectedness with carers (3, 7, 8, 9, 11), and the support of others "in the same boat" with DMD (3, 5, 8, 9, 10).

Vulnerability to prejudice. As the condition of the boys with DMD worsened and they began to use a wheelchair, they felt increasingly noticeable by those around them due to their physical disability (1, 3, 4, 7, 8, 10) and additional equipment: "people like on the street, they kind of freak out when they see the hose" (1). They experienced the prevailing prejudices about people with disabilities, especially the belief that people with physical disabilities must also be mentally impaired (4). As a result, their social relations became more difficult, and establishing a close relationship required significantly more effort of them (3, 6). Adolescents often avoided making new contacts (3) and going out to larger gatherings (8, 10), felt lonely (3, 8), and longed to be seen as a person (3). Various reactions such as anger (1), anxiety (8, 10), or the desire to avoid contact (3, 4, 8) were caused by bullying (3, 8), unethical appeals (3), unwanted attention (1, 8), and an exaggerated desire to help and feeling sorry for them (4, 10).

Risk of isolation. In adolescence, boys are already at higher risk of isolation (3, 4, 8, 10). They tend to communicate less due to the reactions of people around them and (1, 3, 8) bullying (3, 8). Physical and environmental difficulties in reaching certain places and engaging in physical team activities also contribute to greater isolation (1, 3, 4, 10). For some boys, the lack of social contact and communication is filled at a young age by having animals (3) and close ties with relatives (3, 4, 8, 9). Efforts are often made to maintain long-term friendships that have developed since childhood. The risk of isolation may increase in older age when men move out of the house and move away from family, as it can take longer to make new contacts. DMD sufferers are further isolated by a decrease in body mobility, and it is difficult for men to talk about emotional topics and to open up (9, 11). It is therefore important to pay attention to the need for emotional help in the presence of a sense of isolation.

(In)dispensable parental support. Parental support and care were especially important at a young age (3, 4, 8, 9, 10). As DMD-suffering children require more and more intensive care, parents spend significantly more time with them than the parents of non-sick children. Many children spoke about a uniquely close relationship with their mother: she sometimes understood the child's needs even before he would ask for anything (3, 4), and "knows every little detail in his life" (4). It can be difficult for older men with DMD to try to move away from their parents and become more independent. Moving out of a parent's home is often described by them as one of the attributes of an adult, but this separation is often difficult for both the child and the mother (4, 10), who has devoted a long time to care.

Although the relationship with parents is often very close, some children, and older men in particular, said they did not talk to them about painful topics or discuss their concern about the disease (8). Participants tried to protect parents and maintain positivity as much as possible to avoid upsetting them (8, 9). On the other hand, the notion that some parents did not want to talk to their children about sensitive topics either was also mentioned (9). This can leave little room for expressing negative emotions, sadness, fear, and anxiety.

Emotional connectedness with carers. Participants' relationships with carers varied. On the one hand, attempts were made to establish close, long-term relationships with carers because the men were dependent on them to have their needs met (3, 5, 7, 11). On the other hand, a close connection can sometimes make it more difficult to criticize or to apply for more intimate needs (7, 11). One study, where a man wanted to maintain a good relationship but not one that was too close, described "a clear boundary between the volunteers' functional role and the fact that the volunteer is another 'real' person in the room" (11).

Support of others "in the same boat" with DMD. Multiple studies reflected the importance of communicating with others with DMD (3, 5, 8, 9). Other men with DMD may understand and listen because they experience similar difficulties; it can be easier to communicate with them on emotionally intense topics that are too difficult to talk to friends or parents about "because they're in the same boat and they understand" (9). By monitoring older DMD patients, younger sufferers can also learn how to receive support from others (5). On the other hand, in some cases, older people with DMD may frighten younger ones with the severity of their condition, as if showing what awaits them in the future (8). Seen from the other side, people with DMD also tend to provide help and understanding (3) in an attempt to make sense of their lives with the disease.

When dealing with individuals of a similar destiny, men tended to share more intimate and difficult topics, while advice and help from close relatives or health care professionals was viewed quite controversially (3, 10, 11). Men tended to think they know themselves best, so the excessive desire of others to help can have the opposite effect, lowering their self-esteem: "people think they know what they're like…[they say] 'I would wear them if I were like you' but they don't know what it's like to be me" (10).

2.4. Coping processes and resilience

This topic includes 4 subthemes: underestimated strength (1, 3, 4, 6, 7, 9, 11), taking autonomy (4, 5, 7, 9, 11), toward acceptance (1, 4, 5, 8), and active self-management (3, 4, 6, 8).

Underestimated strength. More than one study reflected an underestimation of the resilience of men living with DMD (1, 7, 9). One study found that healthcare professionals avoided talking about difficult topics with men, thus underestimating their ability to break (9). Patients were sometimes treated like children: one man told of how a carer had given him a toy (7). They were also seen as fragile and "breakable," and carers were sometimes overanxious about men's bodies (7). It was also found that "in the delivery of social care, the men reported being treated as largely gender-less if it was set up in such a way that can take away men's independence and autonomy" (7). Meanwhile, boys and men often felt more resilient and stronger than they perceived being valued as (1, 3, 6, 7, 9): "I think we're stronger than they think we are. We deal with lots" (9).

Taking autonomy. As the condition worsens, individuals with DMD seek more autonomy. The opportunity to control surroundings, make significant decisions, and express expectations became one of the strategies of coping with the worsening of the disease (5, 6, 7, 9, 11). Realizing that they could manage their life even if they were in a wheelchair or used a ventilator helped people to come to terms with the current situation and maintain self-confidence. One study described participants' perception that they could obtain bodily autonomy despite being entirely dependent on caregivers (5). Autonomous action led to the acceptance that they could manage their daily activities and some of the things around them.

Toward acceptance. Understanding and accepting the disease occurred gradually and took time (1, 4, 5, 8). One study showed that children initially understood only some aspects of the disease; for example, the need for rehabilitation (4). Doctors must repeat information about the disease and its nature many times so that children remember its essential features (8). Awareness of the disease was strongly influenced by changes in its progression – the weakening of the body, the introduction of wheelchair, and the use of a ventilator (4, 5, 8). At that time, their disease became increasingly real to them, and shock occurred. Participants then faced increasing helplessness: "what triggered my awareness was the fact that I could no longer just get up from the bed, as I had used to" (8). These changes are described as transitional periods, where participants began to face their disease and their habits and routines were adapted to match their new circumstances (1, 4). They started to recognize the progression of the illness and understand what will happen to their bodies (5). Transitional periods were often accompanied by questions about the meaning of the disease (3). Acceptance of the disease, then, occurs by facing and understanding the disease. Accepting the disease seemed to free men from suffering: "you may even prolong your life a bit more if you can accept it and talk about it. You know, if you can get rid of your hurt and anger and depression about it" (9).

Active self-management. Children and adolescents with DMD took more active control of their disease as it progressed. They took actions that depended on them to try to stop the weakening of the body as much as possible (3, 4, 8). Some participants performed a variety of physical exercises at home on their own (3), appreciated rehabilitation and made greater efforts to rehabilitate (8), participated in various physical activities and sports while in a wheelchair (3), and tried to concentrate and focus more on what they could change (3, 4, 6). Participants sought not only to control their physical activity but also their emotional well-being. For some participants, prayer and faith helped them to feel better (2), whilst for others, a conversation with a psychologist (8) or emotional coping exercises (3) were helpful.

Maintaining a positive attitude can be seen as an active strategy for addressing uncertainty and decline (6). Some studies showed that despite the physical limitations, participants still found positive approaches to their experiences (3, 4, 6). "Keeping positive" meant focusing on abilities rather than disabilities and not engaging with thoughts about decline, impairments, and death (6). If they could not change the situation, they tried to change the meaning of the situation (3). On the other hand, seeing the situation as solely positive may lead to an inability to reflect on painful feelings.

2.5. End-of-life experiences

This topic includes 3 subthemes: living in the present (2, 6, 10, 11), experiences of dying (2, 9, 10, 11), and (mis)communication with others about death and dying (8, 9, 10).

Living in the present. People living with DMD said that they thought little about what their future would be. Older participants especially tended to express modest, small desires or expectations for the future (2, 10). Younger respondents expressed hopes for the future such as staying healthy, living independently (for those who did not already), and being in a committed intimate relationship (2, 6). According to one participant, "to focus on his future was to dwell on the 'morbid' uncertainties of the timing of his death" (2). Due to the constant progression of the disease, their future seemed uncertain, but would inevitably lead to deteriorating health and death. Therefore, participants were more likely to live in the present (2, 5, 11) and perform everyday tasks without reflecting too much on future plans.

Experiences of dying. In studies of individuals with DMD, the topic of death is often recurrent. It is emphasized that men talk directly about death very rarely, but more than one study made it clear that death itself is present (2, 9, 10, 11). For example, one study described how residents in an independent living neighborhood experience breathing difficulties, and sometimes choke from mucus entering the ventilator's breathing tube (11). In this case, the machine beeps and urgent assistance is required to remove the secretions. At this point, a person finds themselves close to death, and their life depends only on others. Some men became interested in what death would be like "when you get near to the end" (9). It mattered to them whether it would be painless, or whether they would suffer. Death also changed the life philosophies of older men – they thought less about the future because of fear of seeing death in it (2).

(Mis)communication with others about death and dying. The topic of death is especially relevant for older men with DMD, but, on such a sensitive topic, they rarely tended to talk to others. Although it was often said that such a conversation would be relevant and necessary for them (8, 9), it was quite difficult for them to find someone with whom to do so. For example, men were reluctant to share their thoughts of death with their parents and other relatives (8, 9, 11) to avoid upsetting or intimidating them, and thus protected them by not talking about it. It has been observed that physicians also avoid these topics and talk more about death indirectly; for example, when talking about resuscitation (9). Participants expressed that the person with whom they would like to speak on the subject of death should not only be a passive listener, but should actively engage in conversation, ask questions, have a good understanding of the specifics of DMD, and "be more human" (9): "you want someone with expert knowledge about the condition and what might be involved, and they can answer your questions with authority, but they can also be supportive as well" (9). Men were more likely to talk when they didn't need to initiate the topic themselves, as although they may be unwilling to speak at a certain moment, they would know that they will be heard when the need arises.

3. Discussion

In the last decade, the life expectancy of people living with DMD has increased significantly. As a result, there is a growing need for qualitative research to understand the experiences of these individuals. Some studies aim to analyze the experiences of patients, but they usually focus only on a specific, narrow aspect of research, such as: the transition to adulthood, the experience of one's masculinity, or living with assistive devices. There is still a lack of qualitative research revealing the deep inner psychological world of patients living with this disease. A clearer picture is also absent of their experience of interacting with significant others, interpersonal relationships, and intimacy – particularly when these individuals are known to be at higher risk of isolation. Research shows that questions about the emergence of a new identity in the context of the disease are becoming particularly important.

For those aspects of the psychological state that are highlighted in this study, talking therapy would be an appropriate proposal because all individual needs must be identified and supported. Every DMD case is a very personal journey. The condition psychologically manifests itself in various phenotypes and forms, and develops under the influence of various personal, familial, and social contexts that contribute to the formation of certain stereotypes, attitudes, and values associated with the disease.

It is also worth noting the emotional experiences that boys and young men often tend to experience within themselves without sharing. Research has shown that boys and men often feel better when talking about sensitive topics with other DMD patients who have similar experiences and can understand better what is going on inside. On the other hand, if another patient is in a much more severe condition, it can also frighten younger boys who have not yet fully accepted their illness.

The emotional support provided by others is often received differently. For example, although DMD sufferers' relationships with parents are often much closer than those of non-sick people, boys and older men tend to protect their parents from negative emotions so as not to upset them. Often, parents themselves do not initiate emotionally difficult conversations. The advice of other significant individuals is also not always accepted positively. Men say it is difficult for others to understand what is going on inside the patient because "advisors" do not have the disease.

However, it is essential to understand that it is important for those living with DMD to have the opportunity to speak. The facilitator/listener must be an interested, open, and discerning listener, not only mechanically performing their work.

Conclusion

Qualitative systematic research has provided unique insights into experiences of living with DMD. The results of this analysis should broaden the understanding of how men with DMD perceive and cope with the disease, and the needs and concerns that they encounter. This paper should also help the medical community and policymakers to create a better support system to improve care for men and their families affected by DMD.

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A QUALITATIVE SYSTEMATIC REVIEW OF LIVING WITH DUCHENNE MUSCULAR DYSTROPHY

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Summary

DMD is one of the most severe forms of inherited muscular dystrophies, leading to limited upper limb use in its later stages. Due to medical and technological advances, patients with DMD are living longer and are reaching adolescence and adulthood. It is essential to understand how they perceive and cope with the disease and the needs and concerns that they encounter. The benefits of bringing together qualitative views and studies in a systematic way include gaining a greater breadth of perspectives and a deeper understanding of health, psychological issues, and concerns from the point of view of patients, helping the medical community and policymakers to create a better support system.

Research question: What is the lived-world experience of men with DMD?

Objective: A systematic review of qualitative studies reporting the experience of patients living with DMD in order to identify common themes across all relevant studies.

Methods: The review adopted a systematic search methodology in accordance with PRISMA guidance (Moher et al., 2009) and followed the SPIDER (Sample, Phenomenon of Interest, Design, Evaluation, Research type) search strategy (Cooke et al., 2012) and the analytic procedure for thematic synthesis (Thomas & Harden, 2008). Data sources included: PubMed, Web of Science, EBSCOhost, and Sage Journals. Articles were considered for inclusion if they were qualitative studies written in English that explored the experience of boys or young men living with DMD. The studies were assessed using criteria reported by Thomas and Harden (2008). The quality of the studies was generally high, although several studies were found to be lacking in one or more areas of reporting.

Results and conclusions: 11 studies were included in the review, concerning 132 young people with DMD. Studies were conducted in six countries. The ages of the participants ranged from 10 to 48 years. Consistent with the progression of DMD, all boys and men used wheelchairs for mobility, and 75 received ventilation via tracheotomies or facemasks. Within the papers selected, 5 main topics were identified: experiences of body powerlessness (progressive loss of body control, limited social freedom, lack of privacy), the search for identity (the meaning of becoming an adult man, feelings at the margins, maintaining normality, balancing dependence-independence), challenges in social relations (vulnerability to prejudice, risk of isolation, (in)dispensable parental support, emotional connectedness with carers, support of others "in the same boat" with DMD), coping processes and resilience (underestimated strength, taking autonomy, toward acceptance, active self-management), and end-of-life experiences (living in the present, experiences of dying, (mis) communication with others about death and dying).

Research shows that questions about the emergence of a new identity in the context of DMD are becoming particularly important. The condition psychologically manifests itself in various phenotypes and manifestations, and develops under the influence of various personal, familial, and social contexts that contribute to the formation of certain stereotypes, attitudes, and values associated with the disease. For those aspects of the psychological state that are highlighted in this study, talking therapy would be an appropriate proposal because all individual needs must be identified and supported.

***Keywords**: neuromuscular disease, progressive illness, male gender, qualitative methodology, thematic synthesis.*

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