

Barriers and facilitators to Physical Activity and exercise in Late onset Pompe Disease (LOPD): A qualitative study exploring patient perspectives and experiences

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Abstract

Background: Late onset Pompe Disease (LOPD) is a rare neuromuscular disorder causing progressive muscle weakness. Physiotherapy and exercise may improve general functioning and quality of life in people with LOPD, however, evidence of ways how health care professionals can best support patients to be physically active is sparse. This study aimed at exploring the perspectives and experiences of individuals with LOPD when engaging in physical activity (PA) and exercise.

Methods: Qualitative descriptive study. Semi-structured interviews were conducted with adults diagnosed with LOPD using purposive sampling. Reflexive Thematic Analysis was used to analyse and interpret the data.

Results: 17 adults participated, with a median age of 55 years. Three themes were identified as factors influencing engagement in regular PA and exercise: Perceptions, beliefs, and preferences; State of wellbeing – both physical and psychological; Socio-environmental conditions – access and location, social support, and professional support.

PA was viewed as a positive experience and beneficial for health and wellbeing. Most participants achieved some level of regular PA but often needed motivation to maintain. Resources and support were generally considered lacking, particularly when initially diagnosed.

Conclusion: Engagement in PA and exercise for people living with LOPD is influenced by multiple personal and socio environmental factors; clinicians should use a personalised approach to identify barriers and facilitators and co-develop solutions.

Introduction

Pompe disease is a rare, progressive, neuromuscular disorder (NMD) caused by glycogen accumulation in muscles and nerve cells due to a deficiency of lysosomal enzyme alpha-acid glucosidase (1). Late onset Pompe disease (LOPD) presents after the second year of life, with proximal muscle weakness and respiratory muscle insufficiency being the most prominent symptoms in adults, which can lead to dependency on a wheelchair and requirement of ventilatory support (2).

Additional reported symptoms in individuals with LOPD are pain (3), fatigue (4) and exercise intolerance (5) contributing to difficulties in carrying out activities of daily living (ADL) and reduce physical activity (PA). Individuals with LOPD are shown to be less ambulatory, more sedentary and have higher perceived barriers to becoming physically active than apparently healthy controls (6, 7).

Pharmaceutical treatments, such as Enzyme Replacement Therapy (ERT), have been shown to improve quality of life outcomes, functional capacity parameters and pulmonary function for individuals with LOPD, but their therapeutic efficacy is limited and declines over time (8). Alongside ERT, complementary therapies such as physiotherapy and/or exercise therapy may have an important role in supporting patients' physical functioning and overall wellbeing (9, 10).

A few studies have shown positive outcomes of exercise therapy for adults with LOPD, improving muscle strength, endurance, physical functioning and fatigue (11–15). However, these were small studies, with short follow up periods, involved participants with mild disease severity and using a combination of types of exercise training and adjunct therapies. There has been no qualitative component to these studies exploring individual challenges and acceptability of exercise training, as the focus has been on safety and efficacy of exercise.

Despite the small number of studies, there is a consensus that exercise training, mainly low workload, sub-maximal aerobic training improves general functioning, and quality of life for individuals with LOPD (16). Upholding an active lifestyle could also have positive impacts on physical and mental health, quality of life and prevention of non-communicable diseases for individuals with LOPD, as seen in the general population (17, 18).

Supported exercise training has been the main form of PA studied in LOPD but there are other lifestyle interventions which may help promote PA and decrease sedentary time such as monitoring, advice, and support (19, 20). A retrospective study on adults with LOPD over 10 years found that individuals who remained physically active, meeting the 2010 physical World Health Organisation (WHO) physical activity guidelines, showed better endurance, muscle strength and daily function compared to inactive individuals (21). Participants who continued the structured exercise programme also tended to demonstrate better outcomes in walking distance, respiratory function and activity levels than those that did not participate, suggesting long-term training may offer added benefits.

A small number of qualitative studies involving individuals with disabilities and neurological conditions have identified personal motivation, social support, individual preferences, access to facilities and expert advice as important facilitators of exercise, while symptoms, fear of injury, cost and environmental barriers may limit participation (22–26). More recently, similar factors have been identified in people with slowly progressive neuromuscular diseases (27). However, no qualitative studies have specifically explored experiences of physical activity and exercise in people with LOPD. Understanding these factors is essential to support long-term participation and inform the development of personalised interventions and services.

Research aim

This study aimed to understand the facilitators and barriers to PA and exercise in individuals with LOPD by exploring and describing perspectives, experiences and engagement with PA and exercise.

Methods

Study Design

A qualitative descriptive research design was used for this study on the basis that this type of qualitative enquiry would provide rich descriptive data, from the participants viewpoint, and reveal what influences engagement in PA for people with LOPD and why. A qualitative descriptive approach focuses on the individual's *own* framing around issues and is directed towards the who, what, where and why of events and experiences (28-30).

This study takes a critical realist philosophical position, which assumes that while there may be an objective reality (ontological realism), our experiences and understanding of reality is entirely subjective and socio-culturally shaped, meaning a singular reality or 'truth' is unattainable (epistemological relativism) (31, 32).

This study is reported in accordance with the Consolidated criteria for reporting qualitative research (COREQ) (33).

Ethics

Ethical approval for the research protocol was granted by the University of Birmingham's Science, Technology, Engineering and Mathematics Ethical review committee (ERN_21-1363). Participants provided informed consent through an online consent form. The on-line consent form and demographic information questionnaire were hosted on Online Surveys, a GDPR-compliant and ISO-27001 certified platform.

Interviews were recorded with permission of the participants and transcriptions read through by the researcher to ensure accuracy. Transcripts were anonymised prior to analysing and participants' data storage, confidentiality and anonymity were maintained in accordance with Birmingham University procedures and General Data Protection Regulations 2018.

Interviews have the potential to evoke emotions and unexpected feelings (34). Possible psychological consequences of participating in the study were considered and a distress protocol was developed for participants. The researcher had opportunities for debriefing with co-researchers following the interviews.

Participants and recruitment

Eligible participants were adults (>18years), living in the UK with a confirmed diagnosis of LOPD. There was no requirement of experience with exercise. Participants were excluded if they were non-English speaking or lacked the verbal capacity to participate in an interview.

Participants were recruited by purposive sampling from two UK charities supporting individuals and families affected by LOPD - Association of Glycogen Storage Disease UK and Pompe Support network. Information about the study was distributed to their members as an advertisement received by email in a Newsletter and in members' updates, on social media and on the charities' websites.

The purposive sampling methodology aimed for diversity in self-reported disability, age, gender, time since diagnosis, current exercise activity and whether they are receiving ERT or not. The number of participants was guided by the concept of 'information power', as described by Malterud et al (35), where the more relevant the information the sample holds, the fewer participants are needed. The aim of the study concerns a specific experience e.g. experience with PA and exercise, a purposive sample of participants was recruited holding experiences not previously explored and the researcher had background clinical experience in metabolic disorders, therefore, a sample of seventeen participants was deemed appropriate to ensure quality and adequacy (richness) of the data for addressing the research question, as well as consideration of pragmatic constraints of the research project (35, 36).

Data Collection

Data were collected using semi-structured interviews. A semi-structured interview guide was developed by systematic review and refined by expert opinions from a specialist Inherited Metabolic Disease centre and consultation with patient organisations. There was no participant input or pilot testing due to pragmatic constraints and small participant pool.

Interviews were conducted via the online platform Zoom between February to May 2022. Remote interviewing facilitated inclusion of participants from a wider geographical area for whom travel is difficult. Participants completed the interview at their home, in a private comfortable space and all chose to be unaccompanied. No repeat interviews were performed. Interviews lasted between 50-80 mins, were audio recorded and transcribed. Four of the participants were known to the interviewer before the interview.

Transcriptions were not returned to participants for comments. Field notes were made after each interview to provide a record of observations, impressions, and non-verbal or emotional responses.

Data Analysis

The analytical focus of this study was participants' experiences and perspectives of physical activity (PA) and exercise. A qualitative descriptive design was employed to provide a comprehensive account of these experiences, with data analysed using Braun and Clarke's Reflexive Thematic Analysis (RTA) [31, 38].

RTA offers a flexible and systematic approach to identifying patterns of meaning across a dataset while recognising researcher subjectivity as an integral analytic resource [37]. This approach aligns well with qualitative description, as both seek to remain grounded in participants' accounts and generate rich, clinically meaningful insights without the aim of developing theory. Data analysis followed Braun and Clarke's six-phase, recursive and iterative process [31, 38], with NVivo software (version 12) used to support data coding and organisation.

Results

Participants' Characteristics

18 patients expressed an interest and consented to be interviewed, of whom one (participant 11) subsequently declined prior to interviewing due to technical difficulties, therefore 17 participants (8 male, 9 female) completed the study. The median age of participants was 55 years (range 20-69), 11 (64%) were diagnosed with LOPD more than 10 years ago and 10 (59%) reported exercising more than three times a week. Fourteen participants (82%) were ambulant and eight of those used a gait aid. All participants were accessing a specialist metabolic centre (Cardiff, Salford, Cambridge, Birmingham, and London), with 14 receiving ERT.

Themes

RTA of interview data identified three overarching themes and five subthemes relating to engagement in exercise and PA among individuals with LOPD. Together these themes highlighted the complex interplay of personal, health-related, and environmental factors influencing participation. The three overarching themes were: (1) 'Perceptions, beliefs and preferences influence engagement'; (2) 'State of wellbeing' with the sub themes, 'Symptoms are burdensome' and 'Psychological factors'; and (3) 'Socio-environmental conditions influence access to exercise and PA', with the sub themes, 'Access and location matter', 'Social support is motivating', and 'Professional support is valued'.

Theme 1 - Perceptions, beliefs, and preferences influence engagement

When asked to define what PA and exercise meant to them, most participants could differentiate between PA and exercise. Generally, PA was perceived as everyday activities or movements, such as showering, housework, and exercise was a structured, planned, more 'intense' activity undertaken to improve fitness. However, for those participants with limited physical ability, this terminology was often used interchangeably as daily activities were often described as 'exercise', due to the exertional effort involved.

'I always think exercise is something that you actually make an effort to do, I don't think of the rest as exercise....and now it's difficult' Participant 05

Regardless of each participant's current individual activity level, PA was generally seen as a positive experience, beneficial for one's health and wellbeing, and important in managing some of the symptoms of LOPD. Many also reported it had the potential to make them feel better.

Nearly all participants reported having active lifestyles prior to their diagnosis which generated a more positive attitude about the value of exercise. Beliefs about the impact of PA and exercise, such as improving general health, sleep, maintaining a healthy weight and mood, had a positive influence on PA behaviour. *'I feel like I am doing my heart some good, because I can feel the Cardio'* Participant 08.

The reasons for participating in exercise varied across participants but all stated that exercise was effective in maintaining movement and could help make small gains or possibly slow the deterioration

associated with the disease. This appeared to be a strong incentive to engagement, and participation in exercise was associated with feelings of achievement. Exercise and staying physically active were generally perceived as a personal responsibility and something participants could do to actively contribute to their physical and psychological health *'if you don't use it, you lose it'* Participant 08. One participant described the importance of 'maintaining the connection with your muscles' and how this understanding helped with continuing in regular exercise.

*'it's keeping the mind and the muscles talking, is the key thing. It is not just what you do, it's **why** you do it'* Participant 01

Conversely, one participant described how this attitude can be counterproductive in accepting help and support which could alternately allow greater participation in activity.

'you're often told you should walk, and you have to keep walking to have the ability to walk....that use it or lose it attitude prevents a lot of people from taking advantage of equipment, mobility equipment, wheelchairs.' Participant 09

For participants not currently engaging in exercise there was a divide between those who perceived additional exercise outside of daily activities as no longer beneficial or too difficult and those who were open to exercise as something they *could* do to help themselves.

'Physical activity is my only exercise because I'm so limited in what I can do' Participant 16

For some, a strong motivator was observing how hard it was to re-engage after temporarily stopping exercise or changing a routine. Some shared the belief that they would deteriorate if they hadn't exercised and could see its benefits transferred into making everyday activities easier.

'I'm sure if I hadn't taken on this routine, I would not be getting up so easily' Participant 12

Beliefs regarding their own capabilities were important factors to engagement and some participants described a change in their attitude towards exercise since their diagnosis. An understanding and acceptance of their condition enabled a more positive experience with exercise.

'It's no point saying I can't do that, it's good to try things.....what can I change about the activity to make it fit with the diagnosis rather than what can I change about myself to fit with the activity' Participant 18

For others who were less confident in their capabilities they expressed concerns that there would be negative consequences if they were over-doing an activity or doing an exercise wrong.

'I think there is that fear for people that if they do something and hurt themselves then they've done it wrong' Participant 07

Exercise preferences were frequently based on past activities and experiences. Participants often described exercise activities undertaken prior to their diagnosis and reflected on how they were no

longer able to do what they previously did. Current exercise preferences differed across participants and tended to be associated with confidence in their current physical abilities but also enjoyment, a sense of belonging and convenience were expressed as important in their choices.

'With the use of an e-bike I am the same as everybody else who goes cycling, whereas if I went for a walk, I'd be slower than everyone else.' Participant 18

Some participants demonstrated a knowledge and understanding of their condition and how they used this to determine exercise choice and limits.

'I got diagnosed and learnt about Pompe disease, then I had a much better relationship with exercise and started enjoying it a bit more' Participant 04

Participants valued a wide range of activities, including walking, cycling, swimming, group exercise, technology-assisted exercise, gardening, and household activities, highlighting the importance of meaningful and personally relevant forms of physical activity.

Theme 2 – State of wellbeing

Each participant expressed unique challenges related to their health and well-being which are described within the sub-themes of 'Physical symptoms are burdensome' and 'Psychological factors'.

Physical symptoms are burdensome

All participants commented on the progressive nature of LOPD and described how disease specific physical symptoms such as fatigue, muscle weakness, pain, breathing difficulties, bowel and bladder problems impacted on their ability to participate in exercise and activities, some of which they previously enjoyed. These symptoms impacted on motivation and their perceived physical capacity for exercise.

Fatigue or lack of energy, often linked to muscle weakness and pain, was the most frequently stated barrier to exercise and activity, regardless how long participants had been diagnosed or how physically limited they were. It often impacted on motivation to exercise, limiting their ability or amount they could undertake. Some participants described their experience of fatigue and its impact when trying to engage in PA:

'I think your brain just says, I just can't face that...It's not as though I can't get out the chair....it must be fatigue' Participant 06

All participants described the physical restrictions imposed on everyday activities such as walking, getting up from a chair, or climbing steps and stairs caused by muscle weakness. Each recounted a need to constantly adapt an activity to their physical ability.

'a normal person can whizz a vacuum round very quickly...I have to slow things down and be careful....it doesn't take much for me to be unsettled on my feet' Participant 15

For some participants breathing difficulties were a major barrier as it impacted greatly on daily activities, fatigue, and the type of exercise they could participate in. Six of the participants reported having to use a ventilator to support their breathing and its positive impact on energy levels.

'If your breathing gets compromised the rest of you doesn't function.....I've found it [the ventilator] indispensable because it gives you that charge.' Participant 02

Many participants remarked how every person with LOPD can present differently, with unique needs and abilities. For those early in their diagnosis, aspects of their disability, such as pain and mobility problems were less visible, and often led to false perceptions and judgements on their capability.

Psychological factors

Motivation emerged as a significant barrier to exercise, with over half of the participants described experiencing feelings of inertia or a lack of motivation to exercise, either presently or at some point since their diagnosis. For some this was linked to mood and their mental health,

'..physically exercising, yes it's hard people with Pompe, but not as hard as actually going and getting up and doing those things...I've struggled a lot emotionally and mental health and that side of it...that's had a big impact on exercise for me' Participant 03

Some described themselves as 'lazy', seeing exercise as a chore with limited gains for the effort required. Others expressed frustration and disappointment at the lack of 'gains' made with exercise and observing a decline in activities that they 'could no longer do'. Some described making social comparisons with able-bodied exercisers or their former abled bodied self as de-motivating.

'When you're exercising and can't really do it very well it can be kind of depressing, or to remember you used to be able to do it better, but now can't' Participant 09

Negative exercise experiences often involved professionals not listening and advising exercises which were difficult or impossible to perform, which led to dis-engagement. Some participants described emotional responses such as self-consciousness, embarrassment, loss of confidence in oneself and body and feeling uncomfortable in unfamiliar environments around able-bodied. One participant's response on whether she would consider attending a gym environment was;

'No....I don't want people to look at me and think' what is she doing', because with my disease I'm always judged by 'the look'' Participant 13

Among those participants who were ambulant, due to their muscle weakness, many reported episodes of unsteadiness or experienced falls at home or in public which they described as 'confidence shaking'. Poor balance and anxiety generated from a fear of falls were commonly cited as deterrents to undertaking outdoor activities and participants plan tasks around the anticipation of falling;

'Everywhere I go, I think, how will I get up if I fall. There is no way around this, and I know that people cannot pick me up safely' Participant 17

Other safety concerns were a fear of 'over-doing' an activity and causing further damage to muscles. Many describe a trial-and-error process of finding a balance so as not to over-exert themselves.

'I find that if I over exercise a certain muscle, it just disappears and won't come back for a month' Participant 06

Theme three – Socio-environmental conditions influence access to exercise and PA

Several factors were perceived to help overcome common exercise barriers, described within three sub-themes; Access and location matter; Social support is motivating; Professional support is valued.

Access and location matter

The availability of suitable facilities, access and the environment were described as factors which can facilitate or impeded participation in exercise. Accessibility challenges were frequently encountered by participants, such as inaccessible buildings/pools, unstable or slippery surfaces, presence of stairs, and moving obstacles.

There was a preference towards convenient exercise options and locations for more economic use of their time and not deplete mental and physical energy traveling. Safety concerns were associated with any new environments; unknown locations, new equipment and encountering different people.

'The first thing I would think about is access, is the floor even, are there steps, how packed is the place...all these things go around in your head' Participant 10

Some participants reported limited options for exercise opportunities in their community, with a lack of appropriate facilities, or community based adaptive exercise opportunities. Difficulty accessing continuous support with local rehabilitation services and insufficient local resources were commonly described. Many participants were hesitant about attending traditional fitness centers/gyms where available exercise options may not be tailored to their physical capabilities, along with the excessive costs associated with getting to and attending these facilities. In addition, a perception that staff would lack knowledge on their condition to support them safely and effectively.

'[Fitness instructors] they don't understand what they're actually giving could be detrimental or could even have an impact on muscle growth' Participant 15

This made exercising from home a forced but viable option, with the home environment described as 'enabling' for activity with equipment/aids allowing reasonable independence.

Several participants reported using on-line resources, or wearable devices which they found convenient. However, some participants who trialled them found the often needed adapting for them and that didn't

help their motivation to continue.

'..everyone is so different, one person may be able to do an exercise really easily, I look at that and think I couldn't ever do that, it's sometimes really demoralising even if it's something specific for people with Pompe disease, you look and think, well I can't do that and it's not motivating' Participant 04

Social Support is motivating

Social networks were perceived as the largest facilitator to PA and exercise according to participants. Support was by way of family, friends, co-workers, support groups, health professionals, carers and other patients who offered emotional, physical, or practical support and encouragement. Some participants considered themselves to be self-motivated, but many participants described the benefits of having others motivating them. The additional burden on their carers or family members was acknowledged by those requiring physical support and/or assistance to exercise.

'It's all very well saying 'it's important for you to exercise' but I do think you need the people in your life to help facilitate that' Participant 04

Many participants reflected on the rarity of their condition and valued the connection and support which they had found through on-line Pompe support groups/communities. Other patients' experiences and advice was highly valued and had the potential to provide hope and motivation.

'I value his advice, I see how he looks [referring to another patient] and I see his muscles and I want them too. I want his legs (laugh)' Participant 13

Participants' individual preferences for exercising alone, group exercise or exercise that was also a social activity were also communicated. Exercising with family or friends with shared hobbies was seen as an opportunity for connection and created a sense of normality. Participants described how having others around them, that they could ask for assistance, receive encouragement, often enabled a more structured and disciplined exercise experience.

'if I get up and there's a day that I don't feel like doing it, she'll cajole me into, you know, having a go, 'just do what you can' Participant 14

Professional support is valued

The importance of having access to healthcare professionals with knowledge on LOPD came up repeatedly. Support from knowledgeable health professionals at their specialist centre was seen as fundamental to help self-manage their condition. However, due to the geographical spread of participants many were often a distance away which brought additional travel burdens.

Participants reported being encouraged to remain active within their physical abilities. Many felt that the specialist support, and recommendations they have been given regarding exercise and PA from their treating clinician and/or Physiotherapist were helpful and appropriate.

'It makes such a difference...it helps to be shown what to do, and know that you're doing it right' Participant 05

Although many valued the support, some participants felt that there could be greater emphasis on the benefits of PA. Opportunities for discussions around activity and exercise with clinicians were often too brief or generic rather than individualised, lacking consistency in some cases. However, there was an acknowledgment of the rarity of their condition, variability of individual presentation and how this could be difficult.

*'It's been quite vague and not very consistent.....everyone seems to agree that doing some exercise is useful, it's just that point of **how much** is different.'* Participant 18

Most participants felt it would be beneficial to have access to quality information resources regarding exercise in LOPD, particularly at diagnosis. It was felt education was important, particularly when initiating engagement in exercise, or with any change in physical ability, to set appropriate individualised exercise programme and understand any limitations/precautions.

*'[when diagnosed] these are some good things that you could try to keep your mobility and strength and these things might be detrimental.....education as to what we **can** do.'* Participant 07

Discussion

This study sought to explore and describe the experiences of individuals living with a rare neuromuscular condition, LOPD, when engaging in PA and exercise. Participants reported varied activity levels, with many describing reduced function and decreased engagement in exercise compared with pre-diagnosis.

PA and exercise were often used interchangeably, particularly when daily activities required increased effort. Despite these challenges, all participants viewed PA positively, associating it with improved wellbeing and a sense of responsibility to maintain muscle function, *'if you don't use it, you lose it'*. Although most participants engaged in some form of regular PA, more than half required support to enable them to do so.

Common barriers to PA and exercise reported by participants, including access difficulties, costs, and the need for physical assistance, have previously been described in people with disabilities and neurological conditions (25). Additionally, fatigue, pain, muscle weakness and its impact on mobility, falls, and breathing were described as having the greatest impact on their perceived physical capacity for exercise and motivation. These findings are consistent with those of a qualitative study exploring symptoms and functional impact of LOPD (37) as well as studies in other NMD in which symptom burden, fear of falling, environmental factors and motivation were the most frequent perceived barriers to PA (27, 38, 39). The impact of falls can be far reaching with reduced balance and confidence limiting physical functioning. Fear of falling has been widely recognised as a barrier to PA in older adults and patients with other NMD (27, 40, 41). Falls and balance impairments are frequently reported in LOPD,

they remain underexplored; targeted research could help tailor interventions to manage risk and incidence of falls in this group. Addressing breathing insufficiency, pain, using fall preventative strategies as advised in falls guidelines (42) and implementing fatigue management programmes (43, 44), transferring evidence from similar conditions may help to overcome or manage these barriers.

This study also showed that engaging in exercise can be psychologically and emotionally challenging, as many participants described personal experiences of embarrassment, frustration and worries associated with exercise or exercise environments. This has been observed in other neurological conditions, for example, embarrassment and lack of confidence in the knowledge of professionals' were barriers to participation in community-based exercise (23), and fear of falling/injury, anger and frustration were common emotional responses in multiple sclerosis patients, particularly when PA was perceived to have no positive impact (22).

Lack of motivation and/or difficulty maintaining motivation were among the main reasons for not exercising for the participants in this study. This is consistent with previous evidence identifying motivation as a major barrier to exercise for the general population and patients with neuromuscular conditions (39). Although motivation to exercise can be difficult, a belief in the benefits of exercise and one's abilities ('self-efficacy') has shown to lead to increased participation and motivation among people with other chronic health conditions such as diabetes (45), and arthritis (46). In a study by Anens (38), exploring PA in patients with Charcot-Marie-Tooth (hereditary motor and sensory neuropathy of peripheral nerves), self-efficacy was the most important facilitator for PA alongside enjoyment of the activity.

Self-efficacy is an important feature of Social Cognitive Theory and is a well-known behavioural determinant of PA. It refers to an individual's beliefs in their own capabilities to undertake a specific task and achieve a desired goal and can be influenced by vicarious experiences, past experiences and modelling (47). Although many participants appeared confident in their ability to exercise, maintaining motivation over time with a progressive condition remained challenging. Social Cognitive Theory suggests that healthcare professionals can enhance self-efficacy through strategies such as achievable goal setting, positive feedback, education, peer role models, and ongoing tailored support. Building confidence to exercise safely and consistently may be as important as exercise prescription itself for promoting long-term engagement in physical activity. Similar findings have been reported in people with slowly progressive neuromuscular diseases, where positive beliefs about exercise, social support, and effective self-management strategies supported physical activity participation (27).

Each participant identified many sources of support; family, health professionals, friends, peers, social support groups. Social support was found to be the most important facilitator in a review of qualitative studies on factors influencing participation in PA for patients with neurological conditions (48). Expert knowledge and recommendations on exercise from 'Pompe specialists' and other Pompe patients were highly valued. However, due to rarity of the condition opportunities for group-based exercise or vicarious learning from others with the condition can be scarce. Clinicians in specialist centres may be best

placed to help promote self-efficacy through vicarious dialogue and providing opportunities for patients to share experiences. Also, improved communication between specialist centres and local services can ensure sharing of 'expert knowledge' between professionals and patients.

The value of the participants' knowledge for self-management was evident in this study. Patients with rare disorders often become the expert of their diagnosis (49). Access to evidence-based information and resources supporting PA and exercise in LOPD were seen as important facilitatory factors. However, often environmental, and organisational factors such as limited local accessible facilities, insufficient local resources for supporting activity and lack of access to continuous local services were encountered as barriers. Bandura (50) argues that interventions attempting to change behaviour will be of limited value if they lack the resources and support to be successful. Successful interventions showing sustained increases in PA amongst individuals with neurological conditions include Apps, video coaching, cognitive behavioural therapy, internet-delivered support, and fitness instructors/health professionals to provide support (44, 51–53). These may be low-cost strategies to support long-term participation but further research on efficacy of these interventions would be required specifically for patients with LOPD.

Participants in this study engaged in a diverse range of exercise and recreational activities which suited their interests and physical capabilities, indicating the degree of flexibility required in supporting PA participation. The limited research on exercise in LOPD involve supervised, structured exercise training often in community settings (54), however, structured exercise for some people with LOPD can be hard to access or sustain. Less formalised types of PA and exercise, or a focus on supporting people to be more active using meaningful and enjoyable activities, may be more realistic for long-term engagement and benefit from future research.

Strengths and limitations

This qualitative study provides important findings on the perspectives and experiences of PA and exercise among patients with LOPD. Participants were recruited from specialist clinics across the UK and represented a range of ages, sexes, treatment experiences, and physical activity levels. This diversity enhances the potential transferability of the findings to other adults living with LOPD in similar healthcare settings.

As with most qualitative research, the sample size was relatively small and may not capture the full range of experiences within the LOPD population. However, the study was guided by the principle of information power, whereby participants with diverse and information-rich experiences were recruited to develop a deeper understanding of this complex behaviour rather than to achieve statistical generalisability (35, 55).

Despite purposive sampling, selection bias may have occurred, as individuals with a greater interest in exercise may have been more likely to participate. Measures to minimise potential power imbalance included ensuring informed participation, protecting anonymity and researcher reflexivity (56, 57).

Nevertheless, participants may have been influenced by perceptions of the interviewer as a physiotherapist.

The barriers and facilitators identified were perceived rather than objectively measured, and as such may have limited value as predictors of PA (58). However, consistency with findings from other neuromuscular disease and disability populations strengthens the credibility and potential transferability of these findings.

Clinical implications and directions for future research

Physical activity participation in LOPD is influenced by a range of personal and socio-environmental factors. Clinicians should adopt a personalised approach, working with patients to identify barriers, build on facilitators and support self-management. Specialist multidisciplinary services have an important role in providing tailored advice and promoting long-term engagement in PA.

Health-care professionals should routinely discuss PA during consultations as its recognised benefits for health and wellbeing may help motivate participation. Interventions should focus on key determinants of behaviour, including self-efficacy, fatigue management, and engagement in meaningful activities. Digital health approaches may offer a practical way to support participation and reduce barriers.

Future exercise interventions should be co-designed with patients and incorporate appropriate supervision, flexible delivery, and accessible settings, including home-based options. Given the rarity of LOPD and limited evidence base, future research should prioritise multi-centre, longitudinal, and controlled studies to evaluate effectiveness and sustainability of interventions.

Conclusion

As PA and exercise can be effective self-management strategies for people living with LOPD with the potential to improve health and wellbeing, interventions to promote participation are important. This study indicates that factors that influence participation in PA and exercise for people living with LOPD are complex and require an individualised approach by healthcare professionals.

These findings contribute to our knowledge of factors important for PA behaviour in persons with LOPD and can guide healthcare professionals to support physically active lifestyles for this group of patients.

Although exercise is beneficial in LOPD, high-quality evidence remains limited. Further co-produced, longitudinal intervention studies are needed to support long-term physical activity.

Abbreviations

LOPD

Late Onset Pompe Disease

NMD

Neuromuscular disorder
ERT
Enzyme Replacement Therapy
PA
Physical activity
RTA
Reflexive Thematic Analysis
CMT
Charcot–Marie–Tooth

Declarations

Ethics approval and consent to participate: Ethics approval was granted by University of Birmingham's Science, Technology, Engineering and Mathematics Ethical review committee (ERN_21-1363). Online informed consent was obtained during online screening and again during the interview when interviewer requested consent to audio-record the interview.

Consent for publication: Consent was gained from all participants.

Availability of data and materials: The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request

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