Pompe Disease Monitoring Workshop 2025

Friday 17 and Saturday 18 October 2025
Warwick Conferences,
University of Warwick, Coventry

Highlight Report

Com-NN-UKI-26-00046 June 2026

This meeting was organised and funded by Amicus Therapeutics UK Ltd
in collaboration with an expert faculty who have received honoraria.

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Contents

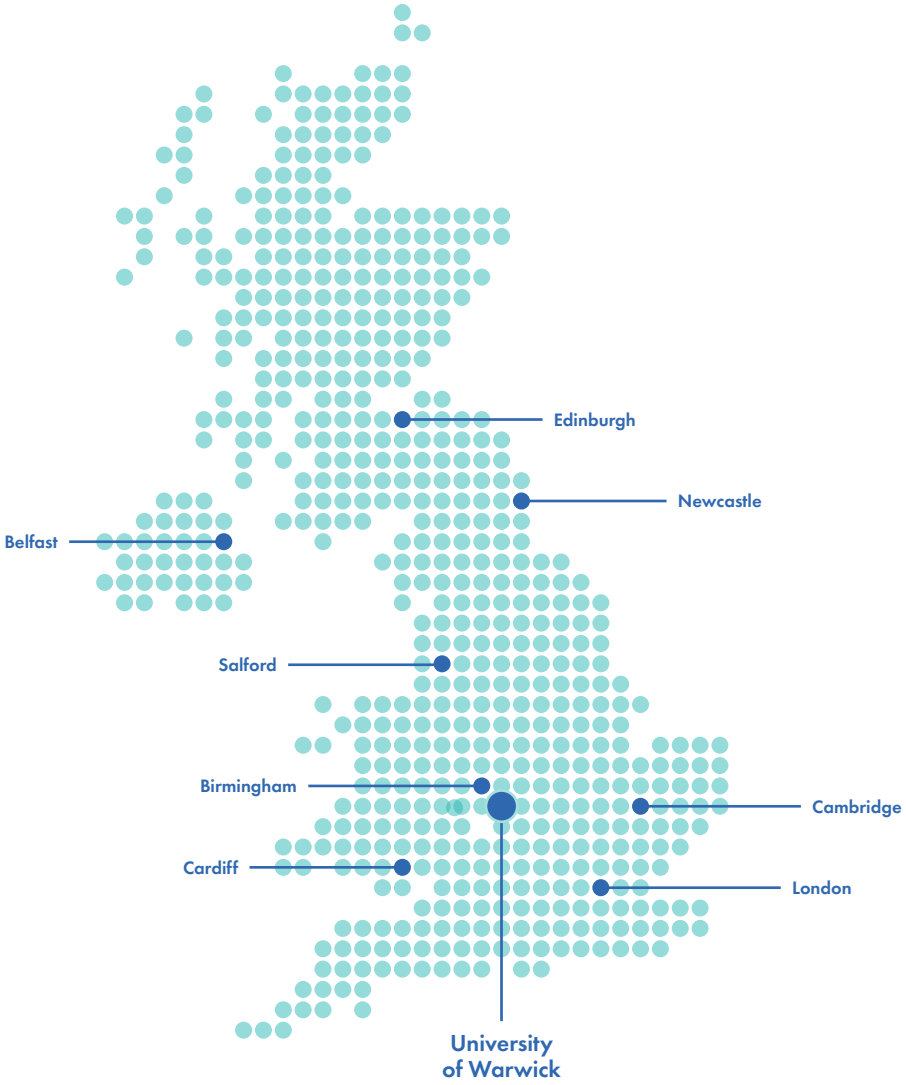
Introduction

The Pompe Disease Monitoring Workshop took place on Friday 17 and Saturday 18 October 2025 at Warwick Conferences, located on the University of Warwick campus. It was funded by Amicus Therapeutics and organised in collaboration with an expert faculty.

Thirty clinicians, physiotherapists and nurses from multidisciplinary teams across the UK came together to share experiences and to discuss the continued implementation of a consistent monitoring approach for patients with late-onset Pompe disease (LOPD).

The workshop focused on the use of patient-reported outcome measures (PROMs) and ventilator data for monitoring, and implementation of the North Star Assessment for limb-girdle type muscular dystrophies (NSAD) in routine clinical practice. Patients and patient organisations discussed their experience and needs for disease monitoring, and representatives from metabolic centres shared their learnings and best practice.

Please consult your meeting workbook to find the resources that were used in the workshop and to which we refer in this report.



Faculty

Chair

Dr Patrick Deegan
Consultant Metabolic Physician
Cambridge University Hospitals
NHS Foundation Trust

Ventilation Lead

Prof Michael Polkey
Consultant Respiratory Physician
Royal Brompton Hospital,
London

PROMs Lead

Ms Liz Morris
Lead Specialist Metabolic Nurse
Cambridge University Hospitals
NHS Foundation Trust

NSAD Lead

Dr Meredith James
Neuromuscular Physiotherapist
John Walton Muscular Dystrophy
Research Centre, Newcastle University

Patient Community Representative

Ms Hülya Apaydin
Chief Executive Officer
Pompe Support Network

With thanks also to the following faculty who helped to develop the content of the workshop:

Ms Ana Amado Fondo
Clinical Nurse Specialist
The National Hospital for Neurology and Neurosurgery, London

Ms Beth Bradshaw
Senior Musculoskeletal Physiotherapist
Cambridge University Hospitals NHS Foundation Trust

Ms Nicola Condon
Clinical Specialist Physiotherapist
University Hospitals Birmingham NHS Foundation Trust

Mr Nigel Cope
Clinical Lead Physiotherapist
Royal Free Hospital, London

Mr Jeff Harvey
Pompe Disease Patient Representative
London

Mrs Amie Lomas
Senior Respiratory Physiologist
University Hospitals Birmingham NHS Foundation Trust

Dr Shyam Madathil
Consultant in Respiratory and Sleep Medicine
University Hospitals Birmingham NHS Foundation Trust

Mr Andrew Oldham
Highly Specialised Metabolic Physiotherapist
Salford Royal NHS Foundation Trust

Dr Azza Osman
Consultant in Chemical Pathology and Metabolic Medicine
University Hospital of Wales, Cardiff

Ms Avtaranpreet Rai
Chief Respiratory Physiologist
University Hospitals Birmingham NHS Foundation Trust

Dr Uma Ramaswami
Consultant Metabolic Physician
Royal Free Hospital, London

Ms Emma Sarrecchia
Metabolic Physiotherapist
Salford Royal NHS Foundation Trust

Ms Shauna Sheridan
Respiratory Physiotherapist
Royal Brompton Hospital, London

Mrs Gemma Stubbings
Pompe Disease Patient Representative
Chelmsford

Agenda

Day 1: Friday 17 October

Time	Session	Speaker/Facilitator
14:00–14:10	Welcome to the Pompe Disease Monitoring Workshop 2025	Dr Patrick Deegan
14:10–14:20	Overview of Pompe disease	Dr Uma Ramaswami
14:20–14:50	Patient experience of Pompe disease monitoring in the UK	Ms Hülya Apaydin, Mr Jeff Harvey, Ms Holly Lumgair and Mrs Gemma Stubbings
14:50–15:00	Update on guidelines for Pompe disease monitoring in the UK	Dr Patrick Deegan
15:00–15:30	Centre experience of Pompe disease monitoring in the UK	Facilitated by Dr Uma Ramaswami Ms Nicola Condon, Mr Nigel Cope and Mr Andrew Oldham
15:30–15:45	*Coffee break*	
15:45–16:15	Ventilation in Pompe disease	Prof Michael Polkey, Mr Jeff Harvey, Dr Shyam Madathil and Ms Shauna Sheridan
16:20–17:50	Practical workshop: ventilation in Pompe disease	Led by Prof Michael Polkey, Dr Shyam Madathil and Ms Shauna Sheridan
17:50–18:00	Day 1 close	Dr Patrick Deegan

Day 2: Saturday 18 October

Time	Session	Speaker/Facilitator
09:00–09:10	Welcome to Day 2	Dr Patrick Deegan
09:10–09:40	PROMs in Pompe disease monitoring	Ms Liz Morris
09:45–10:45	Practical workshop: PROMs in Pompe disease monitoring	Led by Ms Liz Morris, Ms Hülya Apaydin and Mrs Gemma Stubbings
10:45–11:00	*Coffee break*	
11:00–13:00	Centre experience of the NSAD for Pompe disease	Dr Meredith James, Ms Beth Bradshaw Ms Nicola Condon and Mr Andrew Oldham
13:00–13:45	*Lunch break*	
13:45–14:45	Panel discussion	All faculty
14:45–15:00	Next steps and meeting close	Dr Patrick Deegan

Overview of Pompe disease

Dr Uma Ramaswami

Uma Ramaswami is a consultant metabolic physician at the Royal Free Hospital, London. She gave a brief overview of LOPD, highlighting the importance of the multidisciplinary team in the management of patients with the condition.

Uma gave an overview of an international study published in early 2025,[1] which investigated how patients' self-reported LOPD statuses (improving, stable or declining) related to their quality of life as measured by the Short Form (SF)-36 questionnaire. The study revealed three main findings:

1. Self-reported disease progression is associated with a decline in quality of life

'I can't plan for the long term because I don't know how the disease will progress... I need to be afraid of the future.'

'I've been noticing some worsening for a few months. I get more tired, I have some pain that I didn't have before, I lost weight due to lack of appetite.'

'I have accepted that LOPD is a disease that is degenerative and I will slowly deteriorate.'

2. SF-36 questionnaire scores correlate with the reporting of poor health status and 'declining' LOPD status

This suggests that people with LOPD should proactively inform healthcare professionals of their perceived health even if there is no change in standardised test results. Timely intervention may help to maintain physical functioning and general health.

3. The most concerning symptoms for patients with LOPD are:

Muscle weakness and mobility

'I am a prisoner of my body and cannot do what I want to ask of it.'

Pain

'It prevents me from going about my daily life and having a fulfilling social life.'

Respiratory disorders

'The anxiety of not breathing, turning red at the slightest effort and having to gasp just to put on the laces.'

Fatigue

'My whole body hurts from my eyes to my feet.'

The study suggests that the burden of Pompe disease on patients is significant and underestimated. Uma encouraged attendees to speak with the Pompe disease patient representatives at the meeting to gain a deeper understanding of their experiences.

1. Lumgair H, Bashorum L, MacCulloch A, *et al.* Exploring Quality of Life in Adults Living With Late-onset Pompe Disease: A Combined Quantitative and Qualitative Analysis of Patient Perceptions from Australia, France, Italy, and the Netherlands. *J Health Econ Outcomes Res.* 2025;12(1):1–12.

Patient experience of Pompe disease monitoring in the UK

Ms Hülya Apaydin, Mr Jeff Harvey, Ms Holly Lumgair and Mrs Gemma Stubbings

Hülya Apaydin is the Chief Executive Officer of Pompe Support Network, a group of individuals, families, scientists and healthcare professionals aiming to improve the lives of all people living and working with Pompe disease. It is run by members of the Pompe community for the benefit of the Pompe community.

Prior to the workshop, Hülya spoke with six members of the Pompe community to find out more about their views of Pompe disease monitoring in the UK. She presented the main themes as follows:

As three different enzyme replacement therapies are now available, **consistent monitoring of LOPD is more important than ever**. Patients feel that monitoring is just as valuable to them and their families and caregivers, as it is for clinicians, researchers and pharmaceutical companies.

Patients want to be **able to access to their own monitoring data**, particularly to look at their trends over time.

- A digital platform where people could log in to view data without having to contact their clinician would be welcomed by the patient community.

Patients want to **understand what the test result numbers mean**, especially how they translate to the realities of their day-to-day lives.

Patients have **privacy concerns** around their monitoring data – clarity is needed regarding what it is being used for and who has access to it.

Patients are interested in the **utility of artificial intelligence (AI) to identify trends in their monitoring data**. They also want to understand whether AI could be used to identify and flag any issues relating to their condition at an early stage, so that a timely intervention could be implemented. Patients expressed concerns around the accuracy and safety of using AI for these purposes.

Patients are keen to explore the **use of apps** to monitor their own health.

Patients expressed **concerns about inconsistency in care between centres** with one describing it as a 'postcode lottery'.

- In particular, respiratory monitoring was felt to be inconsistent, and not all patients have access to physiotherapists for functional monitoring at their centres.

Hülya handed over to Holly Lumgair, part of the Patient & Professional Advocacy team at Amicus Therapeutics, who introduced two patient representatives who participated at the meeting, and invited them to share their LOPD stories and views on monitoring.

Jeff Harvey lives in London and was diagnosed with LOPD as a teenager following struggles with physical function, such as running and walking up stairs. He explained that he would like to be able to access his monitoring data and potentially use it to inform decisions about his condition.

'I've been completing the Erasmus survey every year since 2003, which includes patient-reported outcome (PRO) questionnaires like the Fatigue Severity Scale. I have also had numerous tests over the years for many things: lung function, strength, bone density, blood tests. I'm disappointed that I can't access the results as I'd like to understand trends over time. Maybe any changes could be linked to events, such as switching treatments or trying a different exercise or diet. This could be very useful in making decisions about my condition in the future.'

Gemma Stubbings is from Chelmsford and was diagnosed with LOPD in her thirties, although she had symptoms from the age of 13. She described her recent annual monitoring assessment, including how useful she found it.

'Monitoring is so important as Pompe disease is highly variable in each individual, and there is no timeline to follow to determine the rate of disease progression. I completed my yearly tests only last week at hospital. I had my regular lung function tests and carried out the NSAD which I found really beneficial. I actually discovered I could complete movements I never knew I could do, and I'll be practicing these daily going forwards. I saw my specialist doctor and I completed my PROMs at home the week before. These had been posted to me, which I much prefer over doing them whilst at the clinic.'

Update on guidelines for Pompe disease monitoring in the UK

Dr Patrick Deegan

Patrick Deegan is a metabolic physician at Cambridge University Hospitals NHS Foundation Trust and led the development of the UK consensus guidelines for LOPD. He explained that these guidelines are now published and can be accessed using this link. During the development of the guidelines the consensus group agreed that the monitoring section should include measures that:

- Reflect patient experience and function
- Cover the full spectrum of LOPD
- Avoid placing excessive demands on patients
- Minimise the time taken to complete assessments
- Enable standardisation across different centres

Patrick gave a brief overview of the monitoring tests that are included in the guidelines:

Required minimal dataset monitoring assessments	Additional recommended options for monitoring
Neuromuscular: motor function	
• **Timed 10-metre walk/run test** in all ambulatory patients to capture disease progression and inform use of mobility aids. • **6MWT** in patients who can safely perform it to facilitate comparison with available clinical trial and retrospective data. • **Appropriate non-ambulatory function tests (e.g. some items in the NSAD)** for those who cannot perform the timed 10-metre walk/run test. • **Systematic recording of the use of a wheelchair** for patients who are non-ambulatory or can walk less than 10 steps.	• **Timed 100-metre or 10-metre walk/run test** should be considered in asymptomatic patients. • **Muscle MRI** should be considered for patients who can comfortably lie flat for 45 minutes. • **NSAD** (personnel training may be required).
Neuromuscular: muscle strength	
• No muscle strength assessments are mandated as part of the minimal dataset.	• **MMT upper and lower limbs (using MRC scale)** should be performed at the discretion of the healthcare team to inform appropriate supportive physiotherapy exercises.
Respiratory function	
• **Recording ventilation use** (yes/no, invasive/non-invasive, overall/daytime number of hours). • **SNIP** in all patients. • **Sitting vital capacity** in all patients. • **Sleep assessment** for patients with SNIP <60 cmH2O, sitting to supine drop in FVC of >20%, or clinical symptoms suggesting sleep apnoea or other sleep disturbance.	• **Supine FVC/FEV_1** should be considered if clinically indicated and the patient can lie down without discomfort. • **PCF** should be considered when clinically indicated. • **Daytime blood gas** should be assessed if clinically indicated.
PROs	
• One Pompe-specific tool and one general PRO tool are recommended to limit burden on patient/clinician. • **R-PAct** scale as a Pompe-specific PRO tool in all patients. • **EQ-5D-5L** as a general PRO tool in all patients.	• **Rotterdam Handicap Scale** should be considered if historical data are available to allow continued monitoring. • **BPI and FSS** may be considered if clinically indicated.

6MWT, six-minute walk test; **BPI**, Brief Pain Inventory; **FEV_1**, forced expiratory volume in 1 second; **FSS**, Fatigue Severity Scale; **FVC**, forced vital capacity; **MMT**, manual muscle testing; **MRC**, Medical Research Council; **MRI**, magnetic resonance imaging; **NSAD**, North Star Assessment for Limb-Girdle type muscular dystrophies; **PCF**, peak cough flow; **PRO**, patient-reported outcome; **R-PAct**, Rasch-Built Pompe-Specific Activity; **SNIP**, sniff nasal inspiratory pressure.

Centre experience of Pompe disease monitoring in the UK

Facilitated by Dr Uma Ramaswami

Ms Nicola Condon, Mr Nigel Cope and Mr Andrew Oldham

Uma Ramaswami introduced the physiotherapist speakers for this session.

Delivery of virtual exercise groups for patients with LOPD

Nicola Condon is a clinical specialist physiotherapist at Queen Elizabeth Hospital in Birmingham and has been running a virtual exercise group for her glycogen storage disease patients since August 2023. Nicola explained how these classes work:

- There is a maximum of six patients per class to ensure participant safety.
- Everyone has an initial virtual consultation before starting to discuss safety and risks.
- The participants are asked to commit to six 45-minute sessions once a week in the first instance.
- There is both a seated class and a standing class to reflect differing abilities.
- Evidence-based exercises are incorporated, which target areas that cause difficulty for patients with LOPD. These cover a spectrum of aerobic, strength, core and balance exercises.
- The classes focus on the joy of movement and celebrate people coming together and doing something active.

Nicola said that the feedback from participants in the classes has been very positive, with many commenting on an improvement in their functional abilities:

'I find it easier to use a self-propelled wheelchair, and can go further distances.'

'I can now lift two heavy dining plates at a time.'

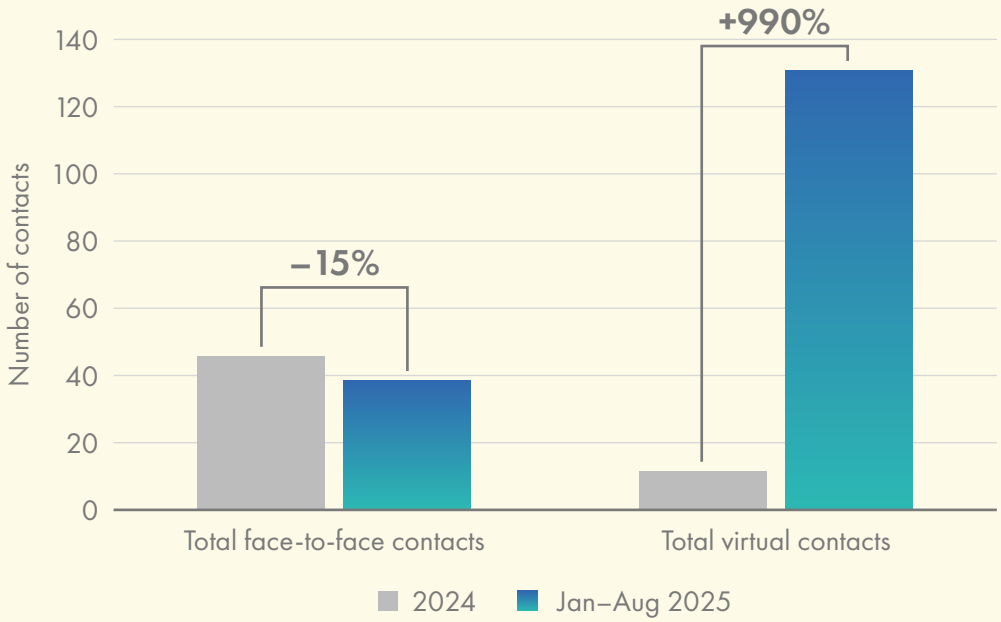
'I managed to lift a cast iron pan above my head into the fridge for the first time.'

Nicola concluded that virtually delivered exercise groups work very well for people with LOPD, and can overcome some barriers to exercise.

LOPD physiotherapy service improvement: monitoring in a specialist metabolic centre

Andrew Oldham is a highly specialised metabolic physiotherapist at Salford Royal NHS Foundation Trust. He explained that a second physiotherapist joined the team in 2024, which provided the capacity to carry out a baseline audit of current LOPD monitoring practices at Salford. Andrew gave an overview of the results of this audit, along with plans to improve physiotherapy care and outcome assessments for patients.

- Having a second physiotherapist on the team facilitated a huge 990% increase in virtual physiotherapy touchpoints from 2024 to 2025. This has mainly been driven by the implementation of a weekly virtual live physiotherapy-led exercise class, as well as the ability to have more regular phone and email contact with patients.

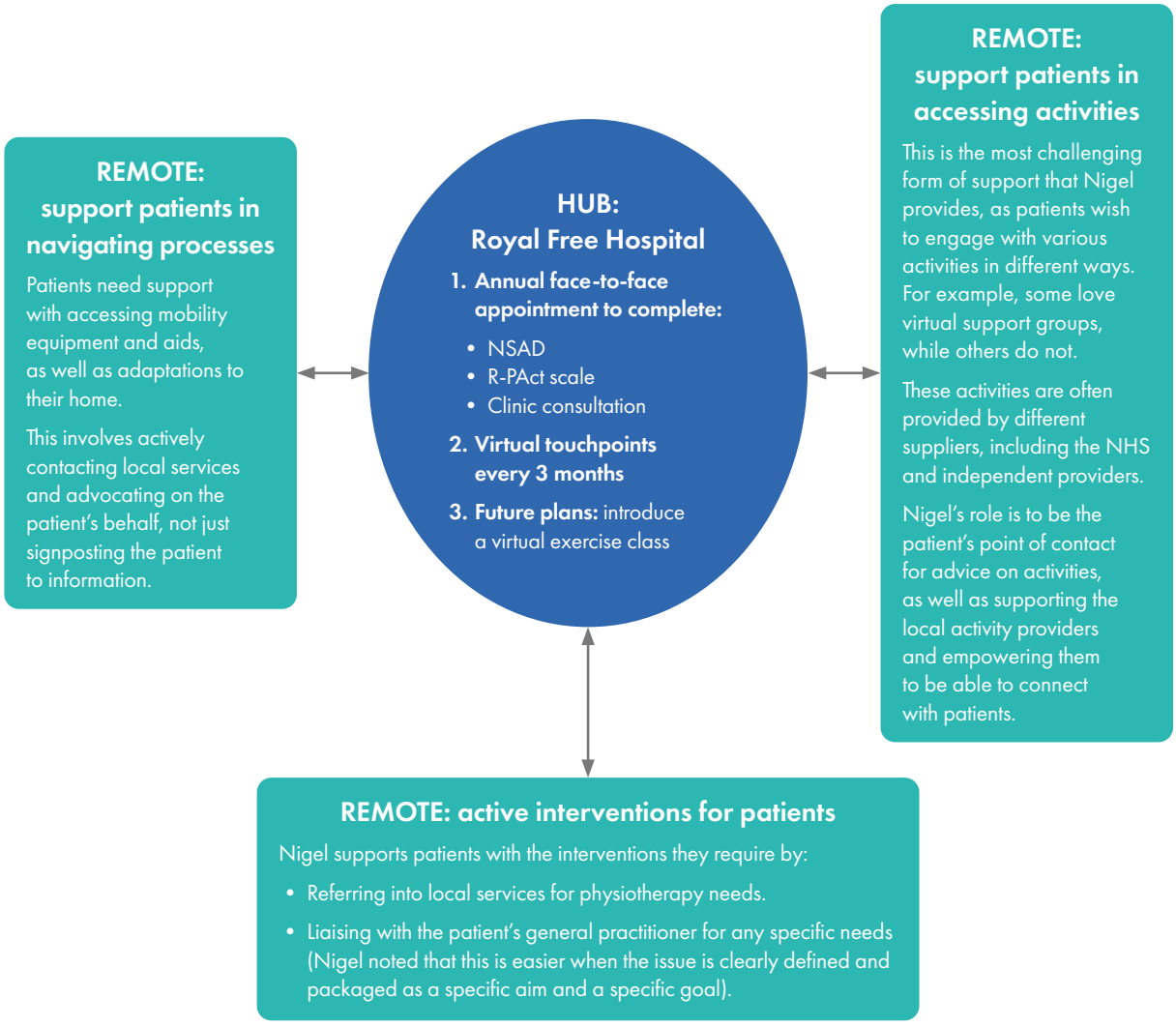


- The physiotherapy team consulted patients with LOPD at Salford to gather their opinions on the PROMs that were used. In terms of patient satisfaction, the Rasch-Built Pompe-Specfic Activity (R-PAct) scale and SF-36 questionnaire scored the highest, but there were only minor differences between all PROMs.
  - Liz Morris has now expanded this project to cover more centres to broaden the findings (see page 21).
- The team has improved physical function monitoring by **introducing the NSAD**, which was completed by 20 patients in the first half of 2025.
- **Regular remote respiratory monitoring** is being explored, as it can be completed over prolonged periods of time, providing a more accurate assessment of lung function and reducing the reliance on in-clinic assessments.

Outreach experience and community support for patients

Nigel Cope is a new clinical lead physiotherapist at the Royal Free Hospital, London, and had only been in post for 3 months at the time of the workshop. He noted that patients often travel long distances to attend London hospitals, so he has been focusing on implementing a 'hub and remote' model of care. This approach involves patients with LOPD receiving centralised care at the Royal Free Hospital, while accessing certain services, such as occupational therapy, through local providers.

To date, Nigel has identified three main areas of remote support that are needed by patients with LOPD. The figure below outlines these areas, along with the role Nigel plays in supporting patients.



NSAD, North Star Assessment for limb-girdle type muscular dystrophies; R-PAct, Rasch-Built Pompe-Specific Activity.

Ventilation in Pompe disease: plenary session

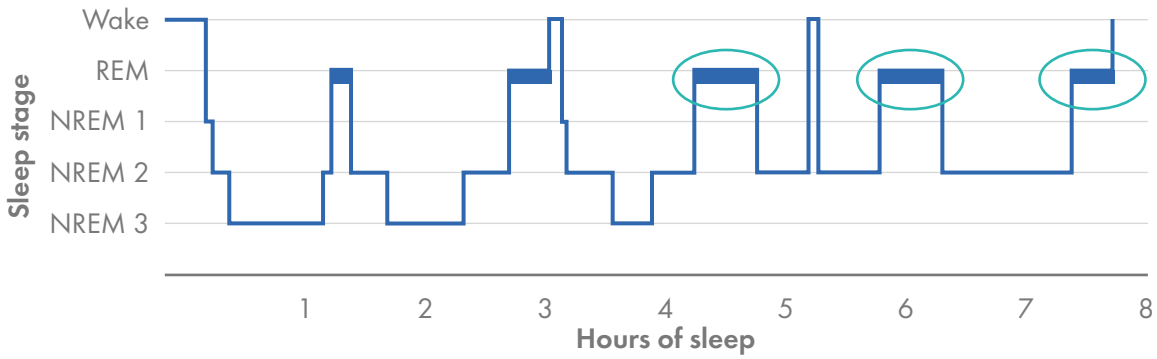
Prof Michael Polkey, Mr Jeff Harvey, Dr Shyam Madathil, Ms Shauna Sheridan and Ms Jude Starkey

Michael Polkey, a consultant respiratory physician at the Royal Brompton Hospital, London, introduced the speakers for this session.

Sleep in late-onset Pompe disease

Dr Shyam Madathil, a consultant in respiratory and sleep medicine at Queen Elizabeth Hospital Birmingham, gave a brief overview of nocturnal respiratory issues in LOPD.

- Respiratory issues often manifest at night because breathing is slightly less efficient than during the day. Muscle tone is especially reduced during the rapid eye movement phases of sleep, which typically occur in the latter half of the night and are circled on the hypnogram below.



NREM, non-rapid eye movement; REM, rapid eye movement.

- Poor sleep quality is common in patients with LOPD. They tend to suffer from obstructive sleep apnoea, nocturnal hypoventilation, and sleep disturbance due to muscle aches and the anxiety/stress of having a progressive disease.
- Poor sleep quality causes frequent awakenings, increased nocturia, waking up unrefreshed, early morning headaches, increased morbidity, and a reduced quality of life.
- At Shyam's centre in Birmingham, 49% (32/65) of patients required nocturnal respiratory support and 65% (11/17) of non-ventilated patients who underwent sleep studies recorded sleep-disordered breathing.

The role of the respiratory physiotherapist

Ms Shauna Sheridan is a respiratory physiotherapist at the Royal Brompton Hospital, London. She discussed the role of respiratory physiotherapists working within a ventilation centre, and summarised their responsibilities as follows:

- Monitor respiratory muscle strength and function, especially through sleep studies, to try and identify any breathing issues at an early stage.
- Support with acclimatisation to non-invasive ventilation, such as troubleshooting mask fittings and teaching cough augmentation techniques to clear mucus effectively.
- Provide acute respiratory support during an exacerbation, including the remote adjustment of ventilation machines; avoiding hospital admissions where possible is beneficial for both the patient and the healthcare teams.

Shauna encouraged metabolic centres to monitor for early signs of respiratory failure during annual reviews and refer patients promptly to ventilation teams. Helpful questions might include asking about the patient's sleep quality, if they wake in the morning with headaches, or whether they have noticed a reduction in their cough strength.

Ms Jude Starkey is a respiratory physiotherapist at Queen Elizabeth Hospital Birmingham and has recently set up a Pompe disease respiratory clinic along with Shyam. The clinic runs one day per month and focuses on assessing secretion clearance and cough augmentation in non-invasive ventilated patients. Jude discussed some of the support she has been able to provide as a respiratory physiotherapist to the 10 patients with LOPD that she has seen so far in the clinic:

Provided individualised treatment plans, looking at breathing techniques and positioning.

Gave lots of advice. For example, kids toothpaste is less foaming than adult toothpaste and so is more suitable for those with salivary secretion issues (as long as it has a suitable fluoride content).

Identified issues and referred patients to appropriate support, such as speech and language therapy.

Solved problems, such as upgrading a patient's NIV machine to allow multiple programmes designed to improve ventilation.

Provided additional monitoring, reassurance and support to patients.

NIV, non-invasive ventilation.

Non-invasive ventilation in LOPD

Michael Polkey gave a brief overview of non-invasive ventilation (NIV) in LOPD, noting that there is a significant and increasing burden to patients as ventilation requirements increase.

Increasing ventilation requirement	Burden to patient
• Night-time NIV only • Typically used for ≥6 hours per night	• Clinic visit and new mask needed twice per year • Maintenance needed once per year
• Advanced NIV • >14 hours per day	• Will need two ventilators • Need frequent sleep studies • Travel needs careful planning, particularly around the length of battery life
• Advanced NIV • >14 hours per day • Expiratory muscle weakness	• May also need to use physiotherapy techniques such as mechanical insufflation-exsufflation (1+ hour per day) • Caregiver may need to provide physiotherapy • May need nebulised saline • May need mouthpiece ventilation • Home visits from outreach team may be required
• Tracheotomy ventilation	• Need a 24-hour skilled carer • Lack of privacy • Travel becomes very difficult

NIV, non-invasive ventilation.

Michael spoke with Jeff Harvey to find out about his experience of ventilation over the course of his condition.

What do you feel is more important in Pompe disease, to preserve strength or breathing ability?

- I think if I answered this question 10 years ago, I probably would have said strength is the best thing to preserve to maintain quality of life. Now that I've lost breathing capacity, I think that I would prefer to preserve that. When I've lost strength, I've always been able to maintain a quality of life through adaptation. But with breathing, there's always the risk of having some kind of crisis that could be deadly.

What are your thoughts on the variability of Pompe disease from one day to another?

- Day to day my breathing is fairly stable, so I do not notice much difference, but once in a while I might feel a bit short of breath and will need to take a few extra double breaths with my ventilator. Sometimes at night my mask can leak, and I might wake up feeling out of breath. I have to adjust my mask or breathe faster to catch up with the breathing. If I have a cold or chest infection, I can get very short of breath and have to do breath stacking with the mouthpiece ventilator to be able to have a good cough. I used to be able to do this with three breaths, but now I can only do two breaths on top of each other.

What hopes do you have for how ventilator data could be used to improve the monitoring of breathing and other aspects of life with Pompe disease?

- It would be useful for me to be able to monitor and understand the data from the ventilator and measure oxygen saturation with a pulse oximeter. It would be especially useful to do sleep studies at home, because going into hospital is very disruptive and you sleep better at home. Collecting data at home might also mean that you could do fewer sleep studies and be able to determine when you do actually need to go into hospital to do one.

Ventilation in Pompe disease: workshop session

This workshop session was divided into three groups, each focusing on a different aspect of ventilation. Group 1 explored ventilator machines and ventilator data, Group 2 focused on cough augmentation, and Group 3 discussed the use of breathing information from patients to inform the development of monitoring tools.

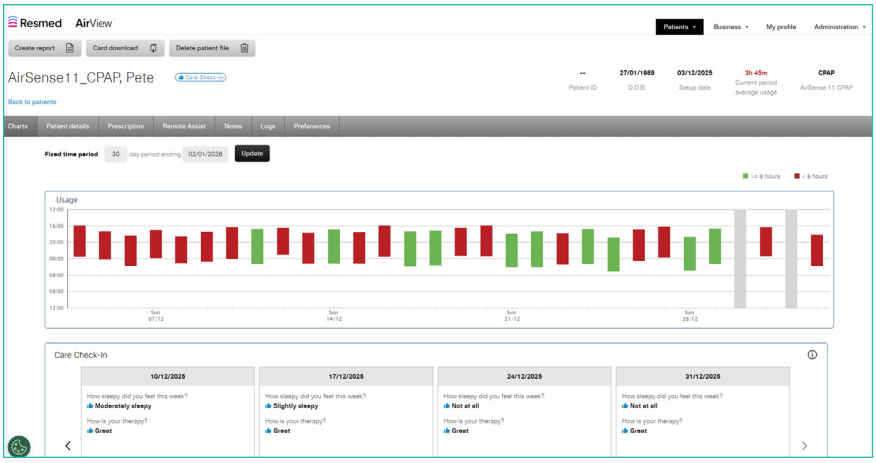
Group 1: ventilators and ventilator data

Group 1 had a demonstration of the Resmed Lumis™ and the Breas Nippy 4™ ventilators, and reviewed ventilator data in relation to the monitoring of patients with LOPD.

- A variety of machines are available to provide NIV.
  - The various ventilators offer different settings, and the choice of ventilator will depend upon patient dependency needs.
  - The patients' ventilators will be set up by the respiratory team at the hospital with fixed inspiratory and expiratory pressures, rather than setting them to deliver fixed volumes of gas. This can compensate better for leaks that occur around the ventilation mask and allows comfortable, effective ventilation for patients.
- There are different masks to suit patient preference: nasal masks, under-nose masks and mouth masks. The mask may be changed for day/night use.
- Some respiratory teams check capillary blood gases to assess a patient's respiratory and metabolic status, to optimise effective NIV therapy. This is much less painful for the patient than doing arterial blood gases.
- NIV devices can be taken as hand luggage on flights along with a letter from the respiratory team.

How to access ventilator data

- Some ventilator models allow clinical teams to remotely monitor their patients. Resmed machines with this functionality use the AirView™ platform, to which all NHS Trusts have access for their patients via a paid subscription.
- The ventilator does not need to be connected to Wi-Fi for remote monitoring to work; it uses phone masts to transmit data.
- The AirView dashboard includes a patient overview and displays their ventilation therapy and compliance data:



How to access ventilator data (continued)

- Patients do not have access to AirView, **but** they do need to consent to data monitoring.
- Individual clinician logins are encrypted.
- Multiple admin accounts can be set up, as well as view-only access options. Respiratory teams can grant permission for metabolic centres to view the data.
- Data can be uploaded in clinic or ahead of the patient's appointment.
- Remote setting changes are available on some ventilator models.

Training videos are available online to help with any questions on the remote monitoring software.

Resmed

Breas

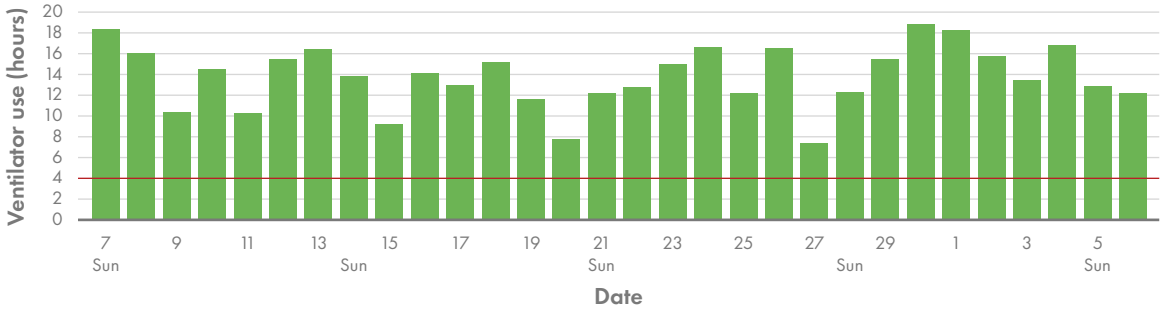
Get in touch with your local respiratory teams to discuss the monitoring of your patients with LOPD on NIV.

Types of ventilator data

1. Hours of use

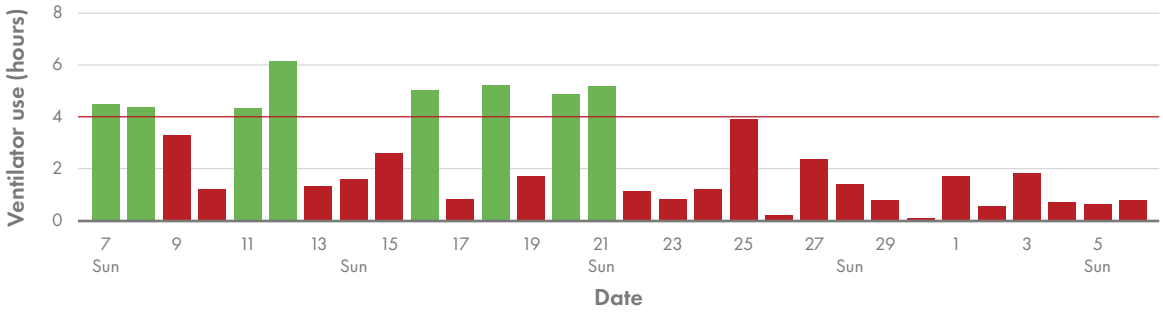
- Tracking the number of hours a patient spends on their ventilator can provide useful headline monitoring data.
- Generally, use of a ventilator for >4 hours per night is considered good compliance.
- If a patient needs to use a ventilator for >14 hours, then this can be a trigger for the provision of a second ventilator.

Good compliance



Ventilator use (hours)	410 hours 51 minutes
Average usage (total days)	13 hours 42 minutes
Average usage (days used)	13 hours 42 minutes
Median usage (days used)	13 hours 43 minutes

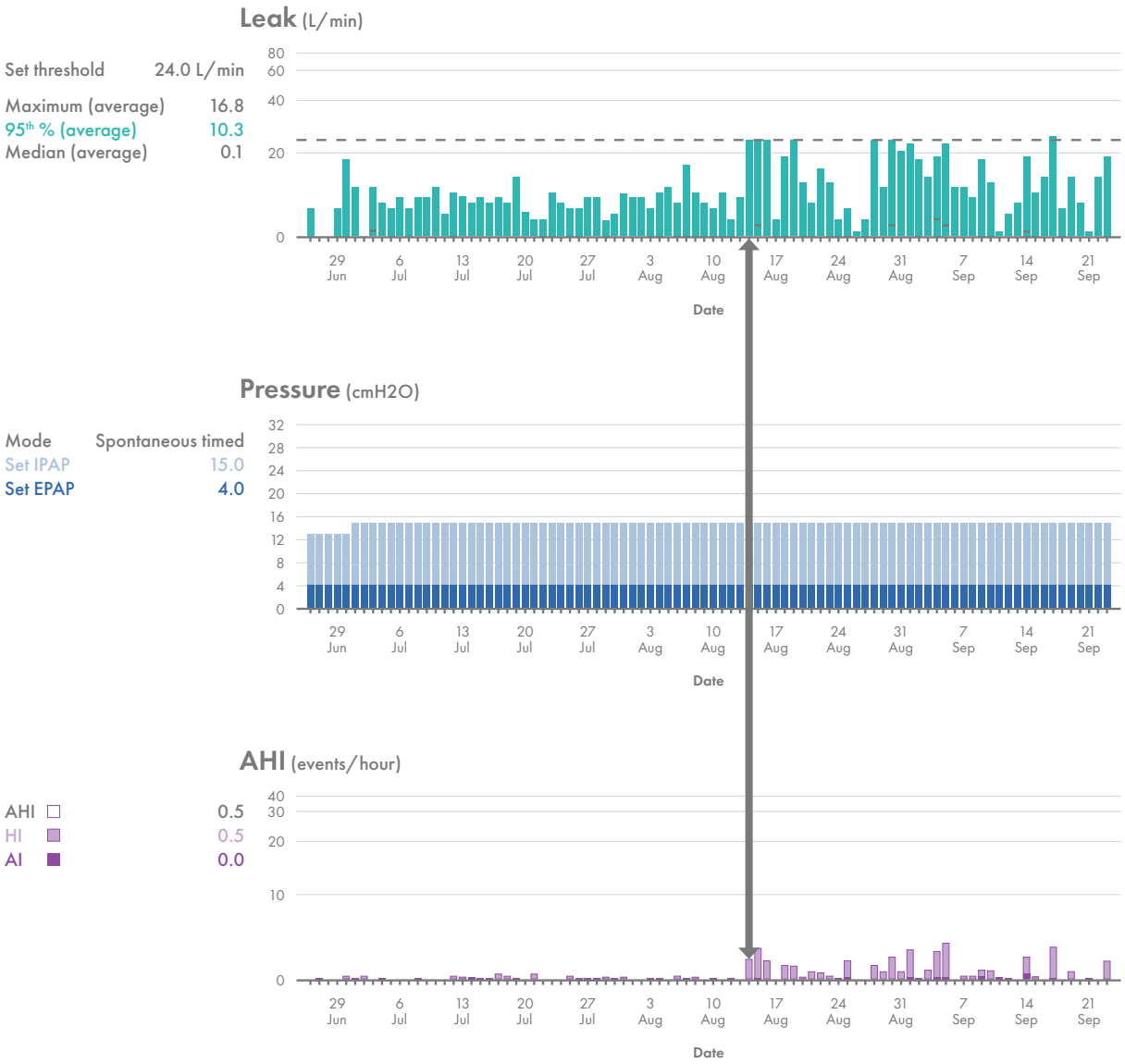
Poor compliance



Ventilator use (hours)	71 hours 2 minutes
Average usage (total days)	2 hours 22 minutes
Average usage (days used)	2 hours 22 minutes
Median usage (days used)	1 hours 42 minutes

2. Pressure and leak

- Ventilator data can also be used to monitor for a leak. A potential leak should be thoroughly investigated; it might just be that the patient removed the mask to use the toilet, or it could be a genuine leak. Check patient compliance and mask comfort, and consider a humidifier or heated tubes to help with mouth and throat dryness to support compliance.
- Most ventilators will compensate for leaks of up to 40 litres, so a small leak is not problematic for patients.



AHI, apnoea–hypopnoea index; AI, airflow index; EPAP, expiratory positive airway pressure; HI, hypopnoea index; IPAP, inspiratory positive airway pressure.

A leak (shown in green) can lead to an increase in sleep disordered breathing (shown in purple). An apnoea–hypopnoea index of up to 5 is considered normal (no sleep apnoea), 5 to 15 is mild sleep apnoea, 16 to 30 is moderate sleep apnoea and >30 is serious sleep apnoea.

Group 2: cough augmentation

Group 2 discussed methods for cough augmentation in patients with LOPD, including a demonstration of the Breas Clearway™ cough assist device and a lung volume recruitment (LVR) bag.

- Peak cough flow gives an indication of cough strength and can be measured by asking the patient to cough into a peak flow meter. These readings are important, and can be used for monitoring, however observing the patient's cough and what they are reporting is equally important.
- The LVR bag can help with inflating the lungs to maximal insufflation capacity, which increases the effectiveness of a cough.
  - The LVR bag can also be used to stretch the lungs and maintain chest wall compliance, which is crucial for the effectiveness of future ventilation.
- Cough effectiveness can also be improved using the breath stacking technique, with or without the help of a ventilator mouthpiece.
  - Breath stacking is breathing in slowly at intervals, stacking one breath on top of the other as tolerance allows, followed by a short hold before slow expiration. This allows the lungs to take in more oxygen than usual and improves cough.[1]
- The Clearway™ device provides mechanical insufflation and exsufflation, the values of which are set and locked by the respiratory physiotherapist. The patient simply has to press 'start'.
  - The mask seal is critical for the effectiveness of the Clearway device, otherwise pressure is lost.

Group 3: using patient information on breathing to develop monitoring tools

Group 3 discussed the aspects of respiratory function that are important to patients. Consideration was given to developing instruments to capture patient breathing and ventilation needs over the course of their disease, and whether this information could serve as a monitoring tool. Key points were as follows:

- Breathing difficulties severely impact quality of life and patients worry about their future ventilation requirements, such as being able to sleep while using a ventilator.
- While adapting to life with a ventilator is a huge psychological adjustment for patients, the use of a ventilator is less of a barrier to going out than going out and struggling to breath without one.
- Ventilator data should be considered in conjunction with qualitative feedback from the patient.
- Some patients do their own sleep studies every night using app-based technology. Continuous data were felt to be more useful than a single time point.
- Hours of ventilator use can provide useful headline monitoring data, can capture data such as oxygen saturation and CO_2 levels, and can prompt conversations around any problematic symptoms such as headaches.
- However, more nuanced data may exist that could be collected to enhance understanding of breathing and ventilation requirements. AI could potentially be used to analyse these aspects within existing datasets.
- The strength of a patient's speech and how long a patient can hold their breath for were proposed as potentially useful monitoring tools.

1. Physiopedia. Breath Stacking. Available from: https://www.physio-pedia.com/Breath_Stacking; accessed December 2025.

Patient-reported outcome measures in Pompe disease monitoring: plenary session

Ms Liz Morris

Liz Morris is the lead specialist metabolic nurse at Addenbrookes Hospital, Cambridge, and is interested in the utility of PROMs to monitor patients with LOPD. Liz gave a brief overview of PROMs as follows:

- A PRO is any outcome related to a patient's health or treatment that is evaluated directly by the patient.
- PROs measure subjective elements of patients' conditions and are recognised by the Food and Drug Administration and the European Medicines Agency as measures of treatment efficacy.
- A key question is how useful PROMs are to healthcare professionals and patients in the real world.

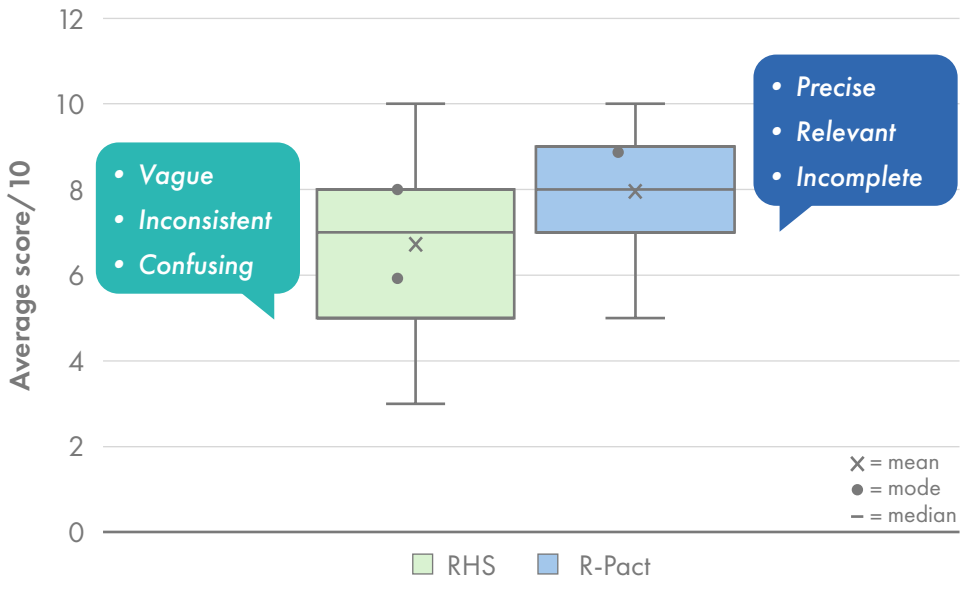
Liz gave an overview of a project that Emma Sarrecchia, part of the physiotherapy team at Salford Royal NHS Foundation Trust, started in 2025. The project sought feedback from patients with LOPD at her centre about the various PROMs that they complete (see page 11). Liz expanded this project to cover more centres to broaden the findings:

- A total of five centres participated and distributed a survey to their patients with LOPD.
- Patients were asked to rate each PROM using a Likert scale (from strongly agree to strongly disagree with four statements), assign an overall score out of 10 for each PROM, and provide any additional comments they wished to share.
- Responses were received from 30 (21%) of the 143 patients who were invited to participate.

Overall results

Disease-specific patient-reported outcome measures

Rotterdam Handicap Scale and Rasch-Built Pompe-Specfic Activity scale

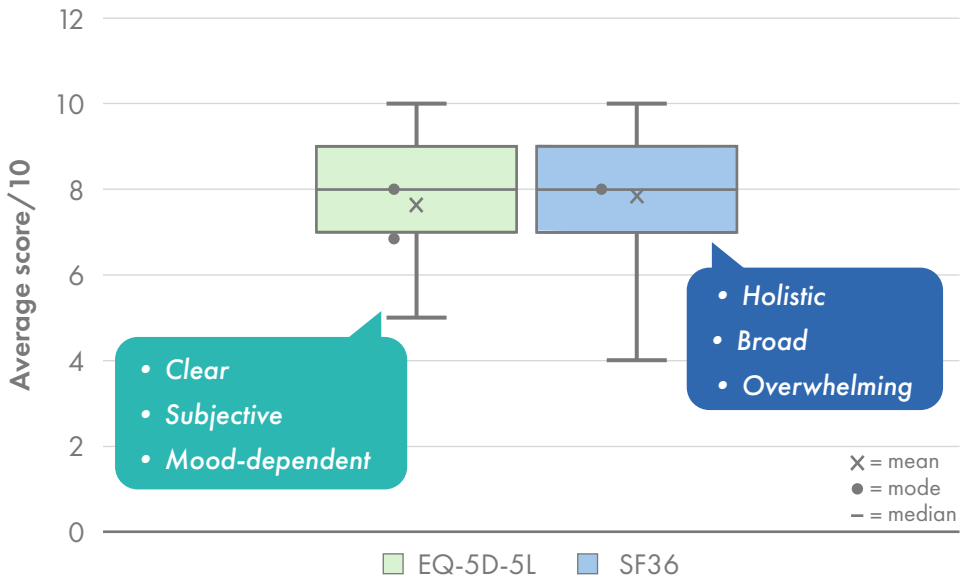


The R-PAct scale performed better than the RHS.

RHS, Rotterdam Handicap Scale; R-PAct, Rasch-Built Pompe-Specfic Activity.

Overall results (continued)

EQ-5D-5L and Short Form-36 questionnaire

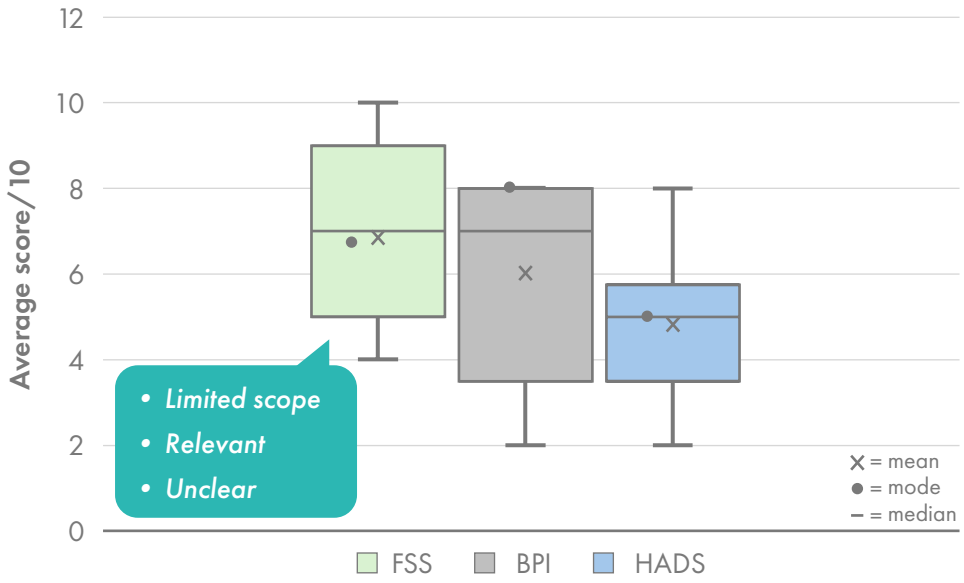


No major difference between these health-related quality-of-life assessments.

SF-36, Short Form-36.

Symptom-specific patient-reported outcome measures

Fatigue Severity Scale, Brief Pain Index, and Hospital Anxiety and Depression Scale



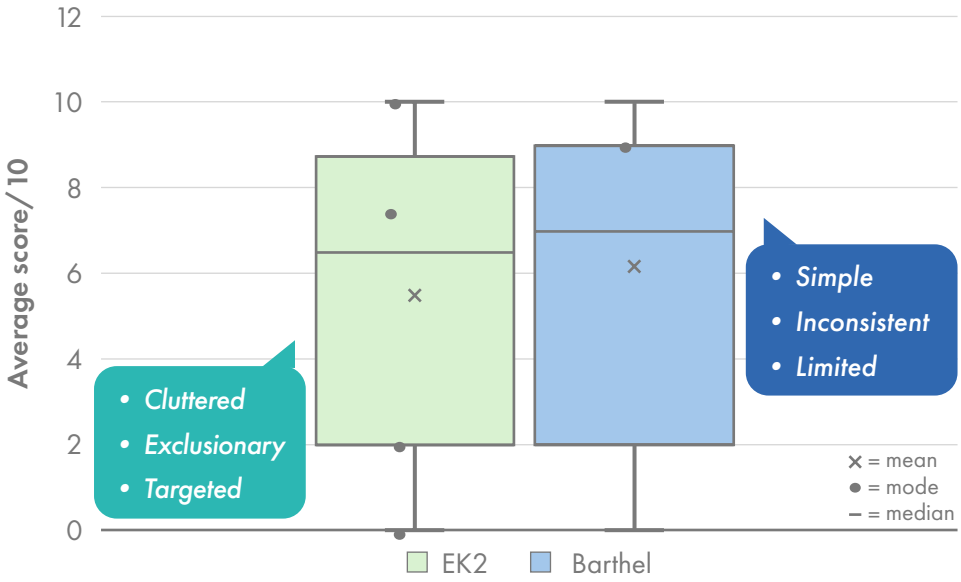
All are relevant assessments for patients who experience these symptoms.

BPI, Brief Pain Index; FSS, Fatigue Severity Scale; HADS, Hospital Anxiety and Depression Scale.

Overall results (continued)

Additional patient-reported outcome measures

Egen Klassifikation Scale Version 2 and Barthel Index



Dichotomous findings; may be useful in patients with decreased mobility when the R-PAct scale is no longer helpful.

EK2, Egen Klassifikation Scale Version 2; R-PAct, Rasch-Built Pompe-Specfic Activity.

Feedback from patients with LOPD

'All the questionnaires ask the same thing in a round about way. Filling in these questionnaires in clinic is time consuming and laborious.'

'Mental health could have more of a focus as this is one of the areas that I believe goes unnoticed.'

'It would be good for patients to be given better insight into what uses the data is put, along with feedback on their data over time.'

'Would be nice to have a QR code or email/fill in online instead of paper. If any patient doesn't want this they should still be able to have a paper copy.'

Future work

Liz plans to work with centres to collect data from more patients and publish the results. She would also like to explore the uses of various questionnaires at different stages of a patient's journey. For example, the R-PAct scale is useful during early stages of LOPD but becomes less relevant as patients become non-ambulatory. The Egen Klassifikation Scale Version 2 (EK2)[1–3] is not applicable for those who are ambulant, but very useful for wheelchair users.

1. Mayhew AG, Eagle M, Steffenson B. S.P.6 Exploratory Rasch analysis of the EK2 scale used in a population of Duchenne muscular dystrophy (DMD). Neuromuscular Disord. 2012;22(9–10):877.
2. Steffensen B, Hyde S, Lyager S, Mattsson E. Validity of the EK scale: a functional assessment of non-ambulatory individuals with Duchenne muscular dystrophy or spinal muscular atrophy. *Physiother Res Int.* 2001;6(3):119–134.
3. Steffensen BF, Lyager S, Werge B, Rahbek J, Mattsson E. Physical capacity in non-ambulatory people with Duchenne muscular dystrophy or spinal muscular atrophy: a longitudinal study. *Dev Med Child Neurol.* 2002;44(9):623–632.

Patient-reported outcome measures in Pompe disease monitoring: workshop session

This workshop session comprised four groups focused on various aspects of PROMs. Group 1 discussed the qualities and attributes of a PROM that are important to healthcare professionals. Groups 2 and 3 carried out PROM assessments and discussions with the patient representatives. Group 4 reviewed and provided feedback on a new patient discussion guide developed by Pompe Support Network.

Group 1: what qualities and attributes of a PROM are important to you as a healthcare professional?

Group 1 discussed the core characteristics and criteria that healthcare professionals consider to be important in a PROM. The discussion was facilitated by Liz Morris and covered the following key points:

What Pompe symptoms are important for you to know about?

Fatigue and pain • Falls • Quality of sleep

Information on mobility, such as whether people need to use furniture to help them move around at home.

Some centres complete a full symptom checklist with patients, which covers breathing, falls, movement, speech, swallowing and mood, amongst other things.

Physician viewpoint: we prefer to understand symptoms that can be treated.

Physiotherapist viewpoint: we are really interested in muscular symptoms and how these affect people in everyday life. Completing PROMs can prompt in-depth conversations with patients about their experiences and needs.

How do you want to obtain the information?

Ideally, PROMs scores should go straight into a patient's hospital record. This has resource implications as someone will need to spend time entering the data.

- Using an iPad to collect PROMs data has worked well at some centres.

Seeing previous scores is sometimes helpful to get a full picture of a patient's progress.

A flexible, hybrid approach may work well. Offer patients the option to complete PROMs on paper or online.

When do you want to know this information?

It is useful to receive scores in advance of a meeting to provide focused conversations in the clinic. This could be facilitated with an app-based system.

Some attendees preferred that patients completed PROMs in the clinic because the answers could then be discussed immediately with the patient.

What do you want to do with this information?

Patients should be given their scores after they have completed the PROMs.

Patients often request to see the PROMs data from their last appointment. However, this can bias their scores on the day if given to them prior to completing their PROM.

What do you think would help patients provide reliable information?

Explaining clearly why PROMs need to be carried out would enable patients to provide reliable information.

Ensuring that patients understand how their data will be used and which aspects of their feedback can be addressed is crucial. It is important not to set unrealistic expectations about what can be achieved.

What would make a PROM acceptable to you?

PROMs need to be:

- Easy for patients to complete
- Representative of the patient's ongoing experience
- Actionable
- Focused on what matters to the patient
- Able to provide more information than can be obtained in a consultation

The information that can be obtained from PROMs must be balanced against the burden to the patient and the healthcare professional of completing multiple questionnaires.

Group 2: PROM assessment deep dive and discussion (R-PAct scale)

Group 2 discussed the R-PAct scale questionnaire with Gemma Stubbings. Key points from this discussion were:

- Both Gemma and the group participants felt that the R-PAct questionnaire was useful, but the discussion highlighted the importance of having in-depth conversations about each response.
  - These discussions can help to clarify functional ability by distinguishing between activities that patients choose not to do, lack confidence in doing, or are physically unable to do.
  - They also add essential context to the patient's responses (e.g. what 'some difficulty' means in real life, and when this might progress to 'a lot of difficulty').
- The physiotherapists were particularly interested in identifying any compensatory strategies that Gemma uses in daily life. Although Gemma reported no difficulty putting on trousers, discussion revealed that this was only possible through significant adaptation (needing to sit down).

Group 3: PROM assessment deep dive and discussion (EK2)

Group 3 discussed the EK2 questionnaire with Jeff Harvey. Key points from this discussion were:

- Most people were unaware of the EK2; only some of the physiotherapists were familiar with it as they were using it routinely for other conditions.
- It is a clinician-led rather than patient-led survey and some of the questions require the clinician to observe the patient for accurate scoring.
- The EK2 is limited to patients who use wheelchairs. However, those who were wheelchair users found it to be a very targeted and helpful questionnaire. Switching from the R-PAct scale to the EK2 may help to better reflect changes in a patient's ambulation and ensure that function can still be monitored in wheelchair users.
- Jeff thought that the EK2 guidance to 'score the best you have done in the last 2 weeks if there is variation between good and bad days' was very helpful to ensure that an accurate picture of overall ability was captured.
- Jeff also noted that some questions could be subject to interpretation. For example, without his mouthpiece ventilator he would struggle to speak to be understood at the back of a large room, but could speak at a decent volume by taking breaths from the ventilator every sentence or two.

Group 4: Pompe Support Network patient discussion guide

Group 4 reviewed the Pompe Support Network patient discussion guide with Hülya Apaydin. The '*Getting ready for appointments at your specialist centre*' section helps patients record symptoms, concerns and questions across relevant topics, so they can use their appointment time at the clinic effectively. The guide was developed in conjunction with a focus group of 15 patients with LOPD who identified the most pressing issues to be included in the guide.

The guide can be accessed here: https://pompe.uk/publications-library/appointment-sheet

Key points from the group discussion were:

- Patients do not usually attend clinic with a list of questions, so the '*Getting ready for appointments at your specialist centre*' section of the guide was felt to be a useful resource. Even if a patient does not make any notes, the headings in the guide are useful prompts for discussion.
- Some centres already use a comprehensive checklist with patients, covering most items in the discussion guide, but it is still useful for patients to reflect before their appointment.
- It would be useful to include boxes in the '*Getting ready for appointments at your specialist centre*' section of the guide to:
  - Record any gastrointestinal symptoms, such as bladder/bowel issues, incontinence and bloating
  - Identify if a patient needs help with accessing benefits or fitness to work assessments
  - List medications that have been started since the last appointment
- If mental health issues are noted in the guide, centres can signpost the patient to Pompe Support Network's psychotherapy service (https://pompe.uk/mental-wellbeing).

Centre experience of the North Star Assessment for limb-girdle type muscular dystrophies for Pompe disease

Dr Meredith James, Ms Beth Bradshaw, Ms Nicola Condon and Mr Andrew Oldham

Meredith James is a neuromuscular physiotherapist at the John Walton Muscular Dystrophy Research Centre at Newcastle University. In collaboration with an international group of physiotherapists, she developed an outcome measure called the North Star Assessment for limb-girdle type muscular dystrophies, which was originally for limb-girdle muscular dystrophy type 2B, but is now validated across all other limb-girdle subtypes.[1]

The pattern of muscle weakness and functional impairments with Pompe disease are very similar to other limb-girdle type muscular dystrophies, and so it was thought that the NSAD could be a useful physical monitoring tool for this disease. Meredith trained attendees at the 2024 Pompe Disease Monitoring Workshop on the use of the NSAD, and three centres are now using it in clinic, with more centres planning to implement it in the near future.

Three physiotherapists from Cambridge, Salford and Birmingham shared their experiences of using the NSAD to assess and monitor their patients with LOPD. Key learnings from the centres has been summarised below.

Case discussion points

- Completing the NSAD highlighted differences between the patient's perception of functionality and demonstrable functional ability. Patients may decline to attempt movements in clinic that they are uncomfortable with as it can be undignified and embarrassing to be lifted off the floor in this environment. It can also be physically uncomfortable or anxiety-inducing for some patients to lie on the floor due to difficulties with breathing. During the assessment, clinical judgement should be used to avoid asking individuals with significant physical impairment to attempt tasks that they are unable to perform.
- Gemma noted that the completing the NSAD has helped her to understand the activities she can perform and has guided her home exercise programme.
- For complex psychological reasons, some patients struggle to accept that more support is needed at home or that changes to everyday life are required to help them with mobility, breathing and safety. Clinics do not want to force people to use equipment, but a better understanding of the challenges a patient is experiencing can aid the physiotherapy team when presenting the available options and explaining the impact of not using them, so that informed choices can be made.

1. James MK, Alfano LN, Muni-Lofra R, *et al.* Validation of the North Star Assessment for Limb-Girdle Type Muscular Dystrophies. *Phys Ther.* 2022;102(10):pzac113.

The **POD-NMD physiotherapy** website contains NSAD resources and training videos that you may find helpful.

Access the site by visiting: https://www.pod-nmd.org/assessment/nsad/

Or scan the QR code:

Panel discussion

Does the order in which the NSAD items are performed matter, and do clinicians really need to continue administering the entire scale once a patient is consistently scoring 0, or could it be shortened to reduce time and patient distress?

The NSAD was originally designed for dysferlinopathy, where calf weakness appears early, so items that are later in the sequence, like walking on tiptoes, are less suitable for Pompe disease where calf weakness is uncommon. Before shortening or modifying the scale for Pompe disease, all items must continue to be tested across patients to understand the true order of functional loss, and identify any irrelevant items. In practice, clinicians already tailor the assessment by omitting items that are clearly impossible based on a patient's initial subjective evaluation.

With newer studies focusing on quantifying glycogen in muscle, when should we start using muscle MRI for monitoring?

MRI methods for assessing muscle glycogen content are still experimental and it is too early for them to be incorporated into routine clinical practice. Some progress has been made in using MRI to monitor the fatty replacement of muscle, but this also presents practical challenges, particularly for patients with respiratory weakness who cannot lie flat for extended periods.

How important is psychological support for patients with LOPD and should this be integrated into Pompe disease services?

Data suggest that clinicians underestimate the psychological burden of Pompe disease, and psychological support is inconsistently available across centres. Some panel members argued that overall, NHS psychology resources should be allocated based on distress rather than being available for all Pompe disease patients. However, clinicians working in Pompe clinics strongly defended the value of having psychologists who were familiar with the condition. Patients often experience denial, loss of function, and emotional strain, especially around diagnosis and disease progression. Centres with embedded psychologists reported clear benefits for patients with LOPD, though the NHS typically funds psychologists for assessments rather than ongoing support. Many patients rely on external networks, such as Pompe Support Network, which offers free counselling services.

Can monitoring data help with practical patient needs like Disability Living Allowance and Personal Independence Payment applications?

Yes. Structured data, such as NSAD scores, can demonstrate patient capabilities, which could support applications for disability benefits.

How should clinicians and patients approach advanced care planning (ACP) in Pompe disease?

ACP discussions are crucial, especially as respiratory support becomes necessary. Clinicians emphasised that patients should decide their own preferences, and the care team's role is to explain what each choice entails. Without ACP, patients entering unfamiliar hospitals risk receiving unwanted interventions. A 'patient passport' could help communicate wishes, milestones and key details. Perspectives on care often evolve over time, so ACP should be revisited periodically. Cultural and personal factors also influence decisions, and there is no substitute for direct conversation with the patient. Without clear direction, healthcare teams or relatives must guess, often leading to unwanted intensive care admissions. Some patients have later requested withdrawal from ventilation – cases which are legally and ethically complex. Therefore, documenting wishes in advance prevents distress and confusion for everyone involved. ACP should cover a spectrum of treatment preferences – not simply 'do everything' or 'do nothing.' It should specify which interventions patients do want (e.g. antibiotics, noninvasive ventilation) and which they do not. The focus should be on safe and appropriate care, not withdrawal of treatment.

How much variation in creatine kinase (CK) levels do you see in patients with Pompe disease compared to other muscular diseases? Do they correlate with disease progression or muscle damage?

In LOPD, CK levels can occasionally be normal. When elevated, they are usually modest (around 500–1000) compared to much higher levels (5000–10,000) seen in Duchenne muscular dystrophy. CK is a marker of muscle disease, but also muscle bulk. It is useful as a safety marker (e.g. if CK levels rise during enzyme replacement therapy, that is concerning), but CK has a poor correlation with disease severity or progression.

What are the challenges of travelling with ventilators and other equipment?

Travelling with ventilators is complex. Some patients carry a backup ventilator and batteries in case of failure. Public transport can be risky for infection, but ventilators have high-quality filters. Air travel needs battery approvals, and emergency equipment, like an artificial manual breathing unit (Ambu®) bag, may be required for situations like MRI scanners.

Conclusion

Thank you all for attending the Pompe Disease Monitoring Workshop 2025! This is the latest in a series of projects aiming to support metabolic centres in the UK to work together to tackle inconsistencies in how LOPD is monitored.

The 2024 Pompe Disease Monitoring Workshop supported multidisciplinary team members from all six centres in England and Wales with practical skills for consistent, patient-centred monitoring. Tools such as the sniff nasal inspiratory pressure (SNIP) test, the physiotherapy-led NSAD, and PROMs were discussed in order to capture what truly matters in daily life for patients with LOPD. Every attendee committed to changing practice, signalling a collective shift toward holistic care.

This latest workshop placed the patient voice at the heart of discussions, with two patients and a patient organisation leader bringing insights gathered from the wider community. A patient-led demonstration of the NSAD, along with case-study experience from three centres, underscored the value of the NSAD in highlighting functional status and guiding conversations about aids, falls and home adaptations. A survey of 30 patients informed decisions on PROMs, and ventilator experts showcased innovations for better capture of respiratory data. Post-event surveys revealed that 90% of delegates intend to adapt their practice to better reflect lived experiences.

We encourage you to keep progressing on the journey you have begun together. Continue collaborating toward your shared goal to improve patient care by standardising and enhancing monitoring. Explore opportunities to publish your work, and do not hesitate to reach out to Amicus if you need any further support.

'It is very rewarding (and possibly even unusual) to see that a small number of efforts made last year to encourage UK centres to collect monitoring data in a more complete and consistent way has led already to improvements in how we work and collaborate with each other. For me, another important take home message is that collecting these data is not only useful for research, but it can have a direct impact on the life of the individual patient, through better communication with healthcare professionals about real-life problems and thus better advice about adaptations and support. I look forward to further progress in showing that these efforts lead to better care for patients.'

Dr Patrick Deegan, PDMW 2025 Chair

Next steps

Guidelines for Pompe disease monitoring in the UK

- Review the guidelines here.
- Contact Patrick Deegan if you would like more information or to discuss the guidelines: patrick.deegan1@nhs.net

Ventilation

- Get in touch with your local respiratory teams to discuss monitoring your patients with LOPD who are on non-invasive ventilation.
- Look out for signs and symptoms of developing respiratory failure at patients' annual reviews so that they can be referred to the ventilation teams at the earliest possible stage.
- Contact Michael Polkey if you would like more information or to discuss using ventilator data for monitoring: mike.polkey@nhs.net

Patient-reported outcome measures

- Support Liz Morris to collect PROMs survey data from more patients and publish the results: liz.morris3@nhs.net
- Explore the use of different PROMs questionnaires at various stages of a patient's journey.

North Star Assessment for limb-girdle type muscular dystrophies

- Continue implementing the NSAD at your centres and share data with Meredith James to allow the assessment to be refined for patients with LOPD: meredith.james@newcastle.ac.uk
- Publish your NSAD case studies.