University Hospitals Birmingham NHS
NHS Foundation Trust

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Final Project Report

Collaborative working agreement between Sanofi and University Hospitals Birmingham to review the Pompe Disease Diagnostic Pathway at University Hospitals Birmingham

Background

The inherited metabolic disorders unit (IMD) based at Queen Elizabeth Hospital Birmingham (University Hospitals NHS Foundation Trust (UHB) provides care and treatment for patients and families affected by inherited metabolic diseases, the unit provides care for over 500 patients. One of the key priorities of the NHS Rare Disease Framework is helping patients to get a faster diagnosis and quicker access to specialist care centres like the IMD unit at UHB.

1.Project Aims and Objectives

To review the diagnostic journey of patients diagnosed with Late-onset Pompe disease (LOPD) referred into Inherited Metabolic Disorders Unit (IMD) Pompe clinic at University Hospital Birmingham (UHB)

2.Project Objectives

Project objectives were:

- Undertake an anonymised review of each patient's diagnostic journey from 1st Pompe related symptom to referral into IMD Pompe clinic at UHB.
- Identify any common gaps and issues that could lead to a delay in diagnosis or suggest areas for improvement.
- To develop an options appraisal of proposed changes to improve the diagnostic patient pathway.

3. Project Outcomes

A patient survey was conducted and sent out by UHB, between January and April 2025 of patients being treated at IMD unit at Queen Elizabeth hospital, as well as some further respondents who were patients contacted via Pompe support network and under the care of other treatment centres in UK.

Key findings:

Time to diagnosis

Patient survey highlighted the extended diagnostic journey undertaken by patients with Pompe disease, as well as the significant impact this can have on mental and emotional well-being.

The average (median) time from first experiencing symptoms that were ultimately attributed to Pompe disease, to receiving a diagnosis of Pompe disease was 8 years, with 45% of patients waiting more than 10 years to obtain a diagnosis.

Factors impacting length of time to diagnosis

In some instances, there was a long delay between patients first developing symptoms and seeking medical attention, with a mean delay of 5 years, highlighting the insidious onset of LOPD and the capability of patients to develop coping strategies around symptoms.

Common barriers to diagnosis included:

- Misattribution of symptoms, with 58% of patients being given an alternate diagnosis before Pompe disease was identified.
- Multiple referrals to different specialities
- Attributing symptoms to life-style factors such as weight gain and exercise.

Common facilitators included:

- Prior connection with Pompe disease – either through a family connection, or seeing healthcare professional with prior experience of Pompe disease
- Early referral to receive appropriate specialist input (e.g. neurology).

Impact on patients

Patients consistently reported a significant impact of their diagnostic journey on their emotional well-being. The greater the delay from symptom onset to diagnosis, the greater the mental and emotional impact was found to be.

On a 1-5 scale of impact on mental well-being, where 5 was “very much” and 1 “not at all”, 76% of patients responded 4 or 5, and for all patients waiting >6 years the average score was 5 (very much).

Themes arising from free-text responses were the anxiety and uncertainty around searching for a diagnosis, the impact of LOPD on daily life and relationships.

Experience with healthcare providers varied but highlighted the importance of multidisciplinary working, particularly physiotherapy input, within the diagnostic process.

4. Timescale

Project commenced Q4 2024
Project completed Q4 2025

5. Resources Utilised

The total cost of the project was: £6,680 This was split into NHS and Sanofi contribution as follows:

Sanofi indirect contribution £2,660
NHS indirect contribution £3,105

Consultancy role during project -Myron Odingo- Health Informatics, Sanofi
Tejiri Osowa- Health Informatics Intern, Sanofi

5. Service impact of project

A report on the outcomes of the review and recommendations based on patient feedback was presented to the IMD team.

This included the following recommendations:

- Create a Pompe disease symptom checklist for primary care physicians
- Education of healthcare professionals on mimic conditions, including need to consider blood tests early in this cohort.

- Enhance awareness of the diagnostic testing process for HCP's and strengthen connections with testing laboratories to improve efficiency and accuracy in Pompe disease diagnosis.
- Educate physiotherapists with training to recognise potential Pompe disease cases, positioning them as crucial early identifiers
- Provide psychological support at diagnosis and throughout disease course.

6.Benefits of Project

The project has been focused on gathering patient perspectives of the process to obtain a diagnosis of Pompe disease. This has developed understanding of:

Patients

- The scale of the challenge in enabling rapid diagnosis of Pompe disease
- Areas to target to improve this, including common misdiagnoses, enabling targeted interventions
- The importance of psychological support given the high impact on patients' mental health and wellbeing

NHS

- The future needs of patients with LOPD to improve their diagnostic journey

Sanofi

- Greater clarity of the patient diagnostic journey to support specific needs of the health professionals within UHB and referring hospitals.
- Improved reputation with relevant NHS referral/ metabolic treatment centres.

7. Recommendations

This work was exploratory, and recommendations have yet to be realised, however actions arising from these have the potential to:

- Speed up time from first presentation with symptoms of Pompe disease to formal diagnosis, with the potential for earlier enzyme replacement therapy.
- Improve healthcare provider awareness of LOPD and the challenges faced by patients
- Provide evidence for increased psychological support to patients with Pompe disease.

The findings will be presented regionally to clinicians with an interest in neuromuscular weakness, with scope to scale nationally subsequently. Where possible, findings will be aimed to be shared more widely through submission for presentation at international conferences or through publication in peer-reviewed journals.

8. Customer feedback

Dr Shayam Madathil, Consultant Physician -Respiratory medicine

"The findings, among other things, highlight the extent of the psychological impact of the delays in reaching a diagnosis on Late Onset Pompe Disease patients. The Sanofi team were very proactive and efficient in getting the project done in a thorough yet timely manner"

Dr S. Quarton, Respiratory registrar

“Working with Sanofi on this project has been a positive experience. Their support in keeping momentum going, and linking to previous similar work was helpful, as was the input with analysis. Where issues or possible inconsistencies were identified during the process these were rapidly addressed, and the result is a report that effectively highlights the challenge in diagnosis of LOPD and helps identify key issues around this for further intervention. This has been an effective team collaboration to reach the result and Sanofi colleagues have been professional and enjoyable to work with throughout”