

Project title	The National Hospital for Neurology and Neurosurgery Pompe physiotherapy service improvement A collaborative working project between Amicus Therapeutics UK Operations and The National Hospital for Neurology and Neurosurgery (The NHNN)
Project rationale	Project Rationale: • Late onset Pompe disease (LOPD) is a rare, progressive disorder affecting muscle and respiratory function due to acid α-glucosidase (GAA) deficiency. • Holistic and consistent monitoring is crucial in a progressive disease such as LOPD to enable better data collection and inform treatment decisions. • Two new ERT medicines are now available, requiring the service to adapt. • The NHNN has no dedicated physiotherapy service within the inherited metabolic service • A novel way of approaching patient regular reviews needs to be investigated
Project objectives	Project objectives • The aim of the project is to review the current Inherited Metabolic Physiotherapy provision and develop a new physiotherapy service in the metabolic service to better coordinate care for Pompe patients. • It will allow all Pompe patients (irrespective of treatment) in the service to access a physiotherapy offer at all stages throughout their disease course, as needed and when any changes occur. • To support Priority 3 of the Rare Disease Framework - to improve the coordination of care for patients with rare diseases. Potential patient Benefits • Improved patient experience of a specialist NHNN physiotherapy service • Improved ability to access NHNN physiotherapy skills and expertise. • To optimise patients' function and accessibility • Improvement in patients' activity levels and or targeted exercise training programs and materials available. • Improvement in patients' quality of life and wellbeing. Potential NHS Benefits • Moving the provision of care from a partly reactive to proactive physiotherapy provision centred around the patient through the use of digital and support care closer to home, with more efficient use of face-to face appointments. • Support the National Rare Disease Framework Priority 3: Better Coordination of Care. • Increased number of data points for more informed clinical decision making regarding starting, switching and stopping of enzyme replacement therapy. • To set up a specialist NHNN Pompe physiotherapy offer • Increased scope of what is possible for future delivery. Potential Amicus Benefits • Partnership working with the NHNN. • Identify challenges and unmet needs in the management of Pompe disease • Improved understanding of the physiotherapy offer and its role in the management in Pompe patients in a large, specialist centre with a diversity of population • As Amicus produces medicines within the Pompe disease area, if overall patient care is optimised there may be an increase in the usage of these products in line with national and local guidelines.
Project period	Q2 2025 – Q2 2027
Financial arrangements	The project will be supported by a pooling of skills and resources from both Amicus and The NHNN, with a direct financial contribution provided by Amicus of £165,916
Plan for publication	Submit preliminary findings to WORLDSymposium 2026 Submission of overall project to WORLDSymposium 2027
Contact details	Laura Clark General Manager Amicus Therapeutics lrclark@amicusrx.com Adrian Capp Therapy Service Manager The National Hospital
Summary of the roles and responsibilities of each party	Roles and responsibilities are agreed and reviewed at monthly steering group meetings