Collaborative Working Project executive summary

Project title	Derriford Hospital Rare Disease Service Review
Partner organisation/s	Sanofi, 410 Thames Valley Park Drive, Reading.RG6 1PT University Hospitals Plymouth NHS Trust, Derriford Hospital, Derriford Road, Plymouth, PL6 8DH
Project rationale	Derriford Hospital is a large teaching hospital in Plymouth, England. The hospital serves Plymouth and nearby areas of Devon and Cornwall. Derriford currently operates an ad hoc system for identifying patients with rare lysosomal storage diseases (LSDs). Currently, there is no clear pathway in place to flag the wide-ranging symptoms these patients present with, which means that patients are often mis-diagnosed, identified late on their diagnostic journey or in many cases not picked up at all. The purpose of the project is to carry out a review of the current journey through secondary care for patients with rare LSDs from first admission/referral to the point of testing for a differential diagnosis. The review will aim to identify opportunities that have been missed in getting these patients a diagnosis earlier and will develop an options appraisal with recommendations to improve the diagnostic pathway. At present, getting a diagnosis can take an average 5-10 years and is the result of a lack of LSD awareness among clinicians and the complexity of the hospital system, being unable to identify them earlier on their diagnostic journey. There are approximately 70 LSDs, of which only some have a treatment available. Therefore, this review will focus on those prioritised by the project lead at Derriford Hospital (approximately six main ones) which may include some for which Sanofi have treatments available. The outcomes of this review (options appraisal) will provide the NHS (National Health Service) with relevant information to improve this service by showing the current inefficiencies to a timely diagnosis.

	To do this requires dedicated project management support, to work alongside the Biochemistry Unit to help co-ordinate and deliver on this service review and evaluation
Project period	Q4 2023 start date Q2 2024 end date
Project objectives	The Project will: - • Identify and review 60 patients tested for the selected Lysosomal Storage Disorders (LSD) in the last 10 years. • Map patient journeys from first presentation into secondary care to Point of testing (POT). • Identify common patterns of missed diagnosis within these journeys. Potential benefits to Patients, NHS and Sanofi are: - **Patient** • Shorten the diagnostic journey for future patients. • Enable more equitable access to care for future patients. **NHS** • Align and deliver on GIRFT (Getting It Right First time) 'a national programme designed to improve the treatment and care of patients through in-depth review of services, benchmarking and presenting a data-driven evidence base to support change.' • Gain a baseline understanding of the current patient journey through an audit of 60 patients tested for some LSDs. • Review of the patient diagnostic pathways will help understand the patient experience and current gaps and inefficiencies in the pathway. • May provide a model for reviewing patient pathways for rare diseases that could benefit other NHS Trusts. **Sanofi** • Greater Clarity of the LSD patient diagnostic journey within Derriford Hospital. • Offers insight into how Sanofi can better support the NHS in 'reducing time to diagnosis 'which is a key priority in the UK Rare Diseases Framework. • Help Sanofi to plan what resources we can offer in the future through education and pathway mapping. • Collaboration and developing partnership with the NHS which builds company reputation.

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