Collaborative Working Project Executive Summary

Project title	The development of an effective identification search query to identify patients with symptoms of Gaucher Disease for review. This will be a collaborative project between Barts and Sanofi known as Project Sirius.
Partner organisation/s	Barts Health NHS Trust Sanofi
Project rationale	Barts Health NHS Trust is an NHS Trust based in London, England. Formed in 2012, the trust is comprised of five hospitals which are located in the City of London and across East London. One of the largest NHS Trusts in England, it has almost 19,000 staff and delivers care to the 2.5 million people of East London and beyond. Around 6,200 patients are treated across Barts Health every day, with an additional 1,500+ people visiting its A&E departments. It provides a huge range of clinical services to its local communities, which include some of the most diverse and socio-economically deprived communities in England. Barts Life Sciences (BLS) is a collaboration between Barts Health NHS Trust and Queen Mary University of London which aims to transform the future of healthcare by supporting innovative research and development of new technologies. The work of BLS covers a broad range of activities from discovery science, product development and testing, to the application of new technology. Gaucher Disease (GD) is a rare Lysosomal Storage Disorder (LSD) that can mimic several haematological conditions and is often misdiagnosed or undiagnosed. Its prevalence is estimated to be 1:100,000, so for our population of 2.5 million, we would expect approximately 25 GD patients within our catchment area. The haematology consultant and data team are looking to address the challenges that people with Gaucher Disease face in seeking a timely diagnosis, treatment, and support. The team consists of clinical and data experts, who will develop a recurring sequential inclusion/exclusion query to flag patients at high-risk of having undiagnosed Gaucher Disease and implement it in the current Trust's system over a period of 12 months. The query will have the capability to run beyond the 12 months the project is projected to run for. The project will address diagnostic challenges related to Gaucher Disease, in line with the 'UK Rare Disease Framework 2021' including misdiagnosis and delays to diagnosis. Currently there is no framework for the identification of people at high risk of developing Gaucher Disease. By year end 2024, a diagnostic search query will be in place to facilitate the diagnosis of Gaucher Disease for Barts Health patients, followed by a referral

	pathway to the haemostasis and thrombosis genomics multidisciplinary team (MDT) and subsequently treatment centre. The aim of this project is to identify patients at high risk of Gaucher Disease with a pathway to enable appropriate follow up. The primary objective of this project is to: 1. Develop a patient identification search query 2. Describe the diagnostic sensitivity and specificity of the search query Sanofi and Barts Health NHS Trust (Barts Health) will partner to deliver the project by combining resources and expertise to: • Develop a sequential inclusion/exclusion query to flag patients with characteristics of Gaucher Disease • Plan the deployment of a recurring query within the Barts Health database/system (Data Core) • Utilise a standard diagnostic pathway following identification of flagged patients for guidance on testing and referral if needed • Progress the project from inception to delivery
Project period	Q4 2023 – Q4 2024
Project objectives	The collaborative working project will deliver the following benefits for patients, the NHS and Sanofi: **<u>Patients</u>** • Reduced diagnostic journey time including an associated improved experience of the system. **<u>NHS</u>** • Less reliance and use of resources employed on disease education of individual health care professionals to make the diagnosis - compared to baseline (previous 12-months data • The potential for further service enhancements of this type through project experience **<u>Sanofi</u>** • Gain insight into the patient diagnostic journey that will enable Sanofi to design future support. • Greater clarity around the development of search queries to aid identification of people with a rare disease so that we can potentially tailor offerings in the future. • As a result of increased patient identification, there may be a potential of the increased use of Sanofi treatments, in line with local and national guidelines.

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