

Executive Summary

A declaration of collaborative working between*

University Hospitals Birmingham NHS Foundation Trust (“UHB”) and Amicus Therapeutics UK Operations Limited (“Amicus”)

UHB and Amicus are engaged in a collaborative working project called:

Fabry Family Screening

This project was initially planned to run from December 2022 to December 2024. The project has been extended and will now run to Q1 2026

Reason for extension

The recruitment and engagement of a genetic counsellor and a genetic practitioner took longer than anticipated and consequently several of the data collection milestones set out in the agreement were not completed in accordance with the original timelines and consequently we have agreed to extend the project.

Project details

The overall aim of the project is to improve the number of individuals who are screened for Fabry disease following the diagnosis of Fabry disease in a family member (the proband).

The project is supported by clinical expertise from University Hospitals Birmingham as well as financial and project management support from Amicus.

It is anticipated that this project will provide information to demonstrate the longer-term importance and value to both patients and UHB of a genetic counselling service, where such a service is implemented in a systematic manner with routine and regular review and audit.

Expected Outcomes

The overall objectives of this project are to support priority one of the Rare Disease Framework - Helping patients get a final diagnosis faster

To improve the number of individuals who are screened for Fabry disease following the diagnosis of Fabry disease in a family member – Leading to earlier diagnosis

Improvement in the coordination of care provided for Fabry patients and their families - Leading to better patient experience

To demonstrate the value of genetic counsellor services to facilitate the improvement in screening uptake

Key Deliverables

To increase the number of Fabry proband family mapping histories recorded;

To increase the number of family members receiving genetic screening

To develop a business case to show the value of genetic counsellors in the Fabry screening service.

To evaluate the project to inform any further developments of the service

